

## **Supplementary information**

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Supplemental Figure S1: PDF, Average base coverage for the target region for case and control samples.

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Supplemental Figure S3: PDF, Coverage of the MIP probe targeting the reported SOD1 c.169+50delTAAACAG variant for all samples.

Supplemental Figure S4: PDF, Histograms of the variants passing and failing the criterion (20 or 30% call rate).

Supplementary Report 1: PDF, overview of all isoforms studied.

Supplementary Report 2: PDF, graphical overview of the performance of each MIP.

Supplementary Report 3: PDF, list of the MIPs not reaching quality criteria.

## Supplementary methods

### Recruitment of cases and controls

Keratoconus cases were recruited during regular clinical practice at the three hospitals mentioned in the manuscript. Keratoconus diagnosis was based on slit-lamp biomicroscopy and videokeratographic evaluation by using the OPD-Scan ARK-10000 (NIDEK, Tokyo, Japan) and ALLEGRO Oculyzer (WAVELIGHT AG, Erlangen, Germany).

Each recruiting center collected a number of controls approximately equal to the number of cases recruited. Controls were not screened for the absence of KC, but this is not necessary due to the low prevalence of KC in the general population. Moskvina et al. have shown that for diseases with a population prevalence in the order of a few percent, including unscreened controls is a cost-effective strategy with little effect on the power.<sup>1</sup> At the prevalence reported by Godefrooij et al.<sup>2</sup>, in the order of 1/375, there might be a few KC cases included in the controls, but the effect on the power is negligible.

### Quality parameters

Initially, 787 patients and 856 ethnically matched controls were included in this study. Based upon **Supplemental Figure S1**, samples not reaching 100x average base coverage across the target region were excluded from the analysis, thus excluding 88 samples from the population (42 cases and 46 controls), resulting in 745 cases and 810 controls.

Quality parameters for including samples and variants, detected using NGS, were fine-tuned by comparing the NGS results – subject to varying quality parameter cutoffs – to Sanger sequencing results, whereby this latter technique was considered to be the gold standard.

Sanger sequencing was carried out in 34 different samples, across different regions of the area targeted by the MIPs. Per sample, between 10 and 409 genomic positions were covered by the Sanger sequencing, adding up to a total of 4013 genomic positions. Starting from these 4013 genomic positions, covered both by the Sanger sequencing and the NGS, the quality parameters for NGS were fine-tuned in a stepwise

way, using the positive predictive value (PPV) as key metric. This value tells the fraction of observed variants (through NGS) that is truly present (according to the Sanger sequencings as gold standard). In other words, if a variant is observed through NGS, we wanted to maximize the probability that this variant is real – which is equivalent to reducing the number of false positives. In addition, we calculated the negative predicted value (NPV), which describes what fraction of the homozygous wild type alleles (according to NGS) is truly homozygous wild type. This is also an indication on the number of false negatives.

**Criterion 1:** Samples reaching at least 100x average base coverage across the target region

		NGS result		Total
		R/R	R/A or A/A	
Sanger result	R/R	3879	83	3962
	R/A or A/A	15	36	51
Total		3894	119	4013

$$PPV = 36/119 = 0.302$$

$$NPV = 3879/3894 = 0.996$$

**Criterion 2:** Criterion 1 + adjusted cutoffs for genotype calling based upon fraction of the alternative allele:

- Alternative allelic fraction below 20% : variant is called homozygous reference
- Alternative allelic fraction between 20% and 80% : variant is called heterozygous
- Alternative allelic fraction above 80% : variant is called homozygous alternative

		NGS result		
		R/R	R/A or A/A	Total
Sanger result	R/R	3928	32	3962
	R/A or A/A	20	33	51
Total		3948	65	4013

$$PPV = 33/65 = 0.507$$

$$NPV = 3928/3948 = 0.996$$

**Criterion 3:** Criterion 2 + excluding positions with depth below 50

		NGS result		
		R/R	R/A or A/A	Total
Sanger result	R/R	2460	9	2469
	R/A or A/A	4	33	37
Total		2464	42	2506

$$PPV = 33/42 = 0.785$$

$$NPV = 2460/2464 = 0.998$$

**Criterion 4:** criterion 3 + excluding variants with a quality by depth below 8

		NGS result		
		R/R	R/A or A/A	Total
Sanger result	R/R	239	1	240
	R/A or A/A	2	33	35
Total		241	34	275

$$PPV = 33/34 = 0.971$$

$$NPV = 239/241 = 0.992$$

Under the quality parameters from criterion 4, if a variant detected by NGS has a 97.1% probability of representing a true signal. Only variants for which a genotype was called in 333 samples (or 20% of samples) were included.



## References

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## Supplementary figure legends:

**Supplemental figure S1.** Average base coverage for the target region for case and control samples.

**Supplemental figure S2.** Predictions of the effect on splicing of the three variants located in the exon-intron boundaries and ranking in the top-15 of single SNP association tests.

**Supplemental figure S3.** Coverage of the MIP probe targeting the reported SOD1 c.169+50\_169+56del variant for all included samples.

**Supplemental figure S4.** Histograms of variants passing and failing the criterion.

**Supplemental table S1.** Overview of genes analyzed in the study and motivation for inclusion.

Gene	Full name	Chr.	Motivation for inclusion
<i>COL12A1</i>	Collagen type XII alpha 1 chain	6	WES
<i>COL2A1</i>	Collagen type II alpha 1 chain	12	WES
<i>COL5A1</i>	Collagen type V alpha 1 chain	9	Literature: associated with CCT <sup>1</sup> , causes EDS <sup>2</sup> , haploinsufficiency leads to thinner corneas <sup>3</sup>
<i>COL5A3</i>	Collagen type V alpha 3 chain	19	WES
<i>DCAF11</i>	DDB1 and CUL4 associated factor 11	14	WES
<i>DOCK1</i>	Dedicator of cytokinesis 1	10	WES
<i>DOCK9</i>	Dedicator of cytokinesis 9	13	Literature: in linked KC locus and variant causes exon skipping <sup>4,5</sup>
<i>EPPK1</i>	Epiplakin 1	8	WES
<i>FASLG</i>	Fas ligand	1	WES
<i>FNDC3B</i>	Fibronectin type III domain containing 3B	3	Literature: associated with CCT <sup>1</sup>
<i>FOXO1</i>	Forkhead box O1	13	Literature: associated with CCT <sup>1</sup>
<i>HLA-G</i>	Major histocompatibility complex, class I, G	6	Literature: expressed in the cornea <sup>6</sup>
<i>IL1RN</i>	Interleukin 1 receptor antagonist	2	Literature: potential disease-causing mutation in KC family <sup>7</sup>
<i>LAMC3</i>	Laminin subunit gamma 3	9	WES
<i>LCA5</i>	Lebercilin	6	WES
<i>P3H4</i>	Prolyl 3-hydroxylase family member 4 (non-enzymatic)	17	WES
<i>LOX</i>	Lysyl oxidase	5	Literature: associated with KC <sup>8</sup>
<i>LOXHD1</i>	Lipoxygenase homology domains 1	18	WES
<i>miR184</i>	MicroRNA 184	15	Literature: mutations cause a spectrum of corneal abnormalities, including KC <sup>9-12</sup>
<i>MMP9</i>	Matrix metalloproteinase 9	20	Literature: increased expression in KC <sup>13</sup>
<i>NLRP1</i>	NLR family pyrin domain containing 1	17	WES
<i>PARP1</i>	Poly(ADP-ribose) polymerase 1	1	WES
<i>PARP2</i>	Poly(ADP-ribose) polymerase 2	14	WES
<i>PRPH2</i>	Peripherin 2	6	WES
<i>RDH13</i>	Retinol dehydrogenase 13	19	WES
<i>SLC4A11</i>	Solute carrier family 4 member 11	20	Literature: potential disease-causing mutation in KC family <sup>7</sup>
<i>SOD1</i>	Superoxide dismutase 1	21	Literature: potential disease-causing mutation <sup>14</sup>
<i>TF</i>	Transferrin	3	WES
<i>TGFBI</i>	Transforming growth factor beta induced	5	Literature: potential disease-causing mutation in KC <sup>15</sup>
<i>TNXB</i>	TENASCIN XB	6	WES
<i>VSX1</i>	Visual system homeobox 1	20	Literature: potential disease-causing mutation in KC <sup>16</sup>
<i>WNT10A</i>	Wnt family member 10A	2	Literature: risk factor for KC <sup>17</sup>
<i>ZEB1</i>	Zinc finger E-box binding homeobox 1	10	WES + literature: potential disease-causing mutations in KC <sup>18</sup>
<i>ZNF469</i>	Zinc finger protein 469	16	WES + literature: causes BCS <sup>19</sup> , associated with CCT <sup>1</sup> , potential disease-causing mutations in KC <sup>20-23</sup>

Chr = chromosome; WES = Whole Exome Sequencing; CCT = Central Corneal Thickness; EDS = Ehlers Danlos Syndrome; KC = Keratoconus; BCS = Brittle Cornea Syndrome.

### **Motivation for inclusion**

Candidate genes labeled as WES were selected based upon a previous whole exome sequencing study on twenty-two families segregating KC (unpublished results). We selected the candidate genes known to be expressed in the cornea, in which we found a variant that segregated – at least partially – with the KC phenotype, and that could possibly affect protein function. We used the following criteria, some of which were different between families with an autosomal dominant (AD) or autosomal recessive (AR) inheritance pattern :

- variants had to be shared across all affected patients from one family and had to be absent in the unaffected family member,
- MAF cutoffs for the rare allele were 0.0007 (for AD families) and  $MAF < 0.05$  (for AR families), according to dbSNP v142, in ESP6500, the 1000 Genomes 2012 release and ExAC version 3,
- synonymous variants (according to RefSeq) were excluded,
- variants had to be located in exons or in regions affecting splicing (including noncoding RNAs),
- in the AR families, the alternative allele had to be present in homozygous state, in heterozygous state for the AD families.

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**Supplemental table S2.** Study population.

		<b>Patients</b>			<b>Total</b>	<b>Controls</b>			<b>Total</b>
		<b>Belgian</b>	<b>French</b>	<b>Italian</b>		<b>Belgian</b>	<b>French</b>	<b>Italian</b>	
<b>Total per ethnicity</b>		251	268	226	<b>745</b>	295	292	223	<b>810</b>
<b>Gender</b>	<b>Male</b>	180	157	141	<b>478</b>	129	109	98	<b>336</b>
	<b>Female</b>	71	99	85	<b>255</b>	166	174	124	<b>464</b>
	<b>Unknown</b>	0	12	0	<b>12</b>	0	9	1	<b>10</b>

**Supplemental table S3.** Overview of exact reference sequences. Hg19 was used as reference genome.

<b>Gene</b>	<b>Reference sequence</b>
<i>COL12A1</i>	NC_000006.11
<i>COL2A1</i>	NC_000012.11
<i>COL5A1</i>	NC_000012.11
<i>COL5A3</i>	NC_000019.9
<i>DCAF11</i>	NC_000014.8
<i>DOCK1</i>	NC_000010.10
<i>DOCK9</i>	NC_000013.10
<i>EPPK1</i>	NC_000008.10
<i>FASLG</i>	NC_000008.10
<i>FNDC3B</i>	NC_000003.11
<i>FOXO1</i>	NC_000013.10
<i>HLA-G</i>	NC_000006.11
<i>IL1RN</i>	NC_000002.11
<i>LAMC3</i>	NC_000009.11
<i>LCA5</i>	NC_000006.11
<i>LOX</i>	NC_000005.9
<i>LOXHD1</i>	NC_000018.9
<i>miR184</i>	NC_000015.9
<i>MMP9</i>	NC_000020.10
<i>NLRP1</i>	NC_000017.10
<i>P3H4</i>	NC_000017.10
<i>PARP1</i>	NC_000001.10
<i>PARP2</i>	NC_000014.8
<i>PRPH2</i>	NC_000006.11
<i>RDH13</i>	NC_000019.9
<i>SLC4A11</i>	NC_000020.10
<i>SOD1</i>	NC_000021.8
<i>TF</i>	NC_000003.11
<i>TGFBI</i>	NC_000005.9
<i>TNXB</i>	NC_000006.11
<i>VSX1</i>	NC_000020.10
<i>WNT10A</i>	NC_000002.11
<i>ZEB1</i>	NC_000010.10
<i>ZNF469</i>	NC_000016.9





	5'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	3'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
HLA-G	Frameshift/nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	nonsynonymous/inframe	0.6127	0.2331	0.1668	NA	0.5724	NA	0.1270	0.1270	0.1219	NA	NA
	synonymous	1.0000	1.0000	0.3357	NA	0.7083	NA	0.7939	0.7939	0.9101	NA	NA
	intronic	0.4276	0.4348	0.3616	0.1209	0.1838	0.3267	0.0310	0.1191	0.0636	0.5524	NA
	5'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	3'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
IL1RN	Frameshift/nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	nonsynonymous/inframe	0.1058	0.1631	0.0690	0.0922	0.0680	0.0582	0.4036	0.7924	0.1628	0.1209	NA
	synonymous	0.0962	0.0525	0.0386	0.0254	0.0404	0.0244	0.2586	0.0238	0.2228	0.0540	NA
	intronic	0.2701	0.2701	0.0992	0.1038	0.0888	0.0888	0.1150	0.1150	0.1858	0.2068	NA
	5'UTR	0.4750	NA	NA	NA	NA	NA	0.1843	NA	NA	NA	NA
	3'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	
LAMC3	Frameshift/nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	nonsynonymous/inframe	0.6167	0.2261	0.4406	0.1459	0.1868	0.1598	1.0000	0.9123	0.9960	0.7213	NA
	synonymous	1.0000	0.1858	0.4775	0.0916	0.6573	0.1259	0.3978	0.1419	0.6693	0.4206	NA
	intronic	0.7601	0.1714	0.3766	0.0846	0.5514	0.1309	0.4942	0.3837	0.6144	0.4426	NA
	5'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	3'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	
LCA5	Frameshift/nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	nonsynonymous/inframe	0.1058	0.1058	0.0288	0.0338	0.0422	0.0382	0.0253	0.0253	0.1938	0.1938	NA
	synonymous	1.0000	1.0000	0.5914	0.6334	0.4735	0.4765	0.7707	0.7707	0.7393	0.7463	NA
	intronic	0.0674	1.0000	0.7722	0.6074	0.7862	0.5974	0.0229	0.5559	0.0286	0.8591	NA
	5'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	3'UTR	1.0000	1.0000	NA	NA	NA	NA	0.7984	0.7984	NA	NA	NA
LOX	Frameshift/nonsense	1.0000	1.0000	NA	NA	NA	NA	0.5755	0.5755	NA	NA	NA
	nonsynonymous/inframe	0.1636	0.1636	0.0500	0.0548	0.0770	0.0738	0.7398	0.7398	0.1174	0.1249	NA
	synonymous	0.6258	0.6258	0.7912	0.7902	0.7732	0.7542	0.8931	0.8931	0.5025	0.5075	NA
	intronic	1.0000	1.0000	0.6893	0.6773	0.6833	0.6873	0.6628	0.6628	0.9031	0.9171	NA
	5'UTR	0.6257	0.6257	0.7473	0.7582	0.7942	0.7892	0.7749	0.7749	0.4995	0.5115	NA
	3'UTR	0.5006	0.5006	0.8801	0.8561	0.8801	0.8661	0.8754	0.8754	0.3836	0.3516	NA
LOXHD1	Frameshift/nonsense	0.7447	0.6751	0.4266	0.3197	0.4006	0.2897	0.7738	0.3183	0.8162	0.5135	NA
	nonsynonymous/inframe	0.8002	0.2399	0.7343	0.1146	0.3526	0.1968	0.8336	0.6360	0.6953	0.4206	NA
	synonymous	0.9196	0.1040	0.5135	0.0614	0.2927	0.0746	0.5914	0.3967	0.6853	0.5155	NA
	intronic	0.5959	0.4796	0.7762	0.2687	0.8182	0.3776	0.9728	0.7452	0.9950	0.9421	NA
	5'UTR	0.6239	NA	NA	NA	NA	NA	0.3940	NA	NA	NA	NA
	3'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	
miR184	ncRNA_exonic	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	ncRNA_upstream	0.2330	0.2330	NA	NA	NA	NA	0.1268	0.1268	NA	NA	NA
	ncRNA_downstream	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
MMP9	Frameshift/nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	nonsynonymous/inframe	1.0000	0.0954	0.3127	0.0386	0.6394	0.0654	0.8086	0.7965	0.2797	0.0908	NA
	synonymous	0.3549	0.3549	0.1359	0.1374	0.1798	0.1838	0.0520	0.0520	0.2557	0.2567	NA
	intronic	0.0067	0.0312	0.0014	0.0046	0.0110	0.0240	0.1082	0.0285	0.0762	0.0082	NA
	5'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	3'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	
NLRP1	Frameshift/nonsense	0.4844	0.4844	NA	NA	NA	NA	0.2094	0.2094	NA	NA	NA
	nonsynonymous/inframe	0.6183	0.6604	0.8771	0.5205	0.7872	0.4885	0.8444	0.8629	0.6054	0.6873	NA
	synonymous	0.7219	0.8084	0.7103	0.3756	0.8551	0.4525	0.4648	0.0633	0.3836	0.1389	NA
	intronic	0.8752	0.8182	0.7393	0.4326	0.6184	0.3696	1.0000	0.8925	0.9910	0.9311	NA
	5'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	3'UTR	0.4348	0.4348	0.1289	0.1269	0.5355	0.5335	0.1241	0.1241	0.1548	0.1528	NA
P3H4	Frameshift/nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	nonsynonymous/inframe	0.5100	1.0000	0.7972	0.5964	0.7223	0.4496	0.7686	0.7686	0.4326	0.8521	NA
	synonymous	0.0870	0.0598	0.0684	0.0396	0.0250	0.0156	0.3392	0.7765	0.2048	0.0270	NA
	intronic	0.4910	0.3541	0.2488	0.1578	0.1948	0.1668	0.7006	0.4300	0.6763	0.3866	NA
	5'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	3'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	
PARP1	Frameshift/nonsense	0.2508	0.2508	0.9361	0.9481	0.9311	0.9231	0.9000	0.9000	0.1568	0.1638	NA
	nonsynonymous/inframe	1.0000	0.2930	0.5115	0.1459	0.2897	0.1249	0.7133	0.8892	0.5674	0.2028	NA
	synonymous	0.0591	0.5838	0.8881	0.7742	0.8941	0.7792	0.0712	0.8664	0.0448	0.1153	NA
	intronic	1.0000	0.4580	0.4206	0.2418	0.6723	0.3676	0.1804	0.2769	0.3596	0.5335	NA
	5'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	3'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	
PARP2	Frameshift/nonsense	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
	nonsynonymous/inframe	0.1997	0.0350	0.0430	0.0072	0.2737	0.0294	0.0241	0.0043	0.1678	0.1234	NA
	synonymous	0.0962	0.0962	0.0538	0.0514	0.0350	0.0442	0.6529	0.6529	0.1014	0.0888	NA



5'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
3'UTR	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA

MAF = Minor Allele Frequency; Vt = Variable Thresholds methods; kbac = kernel-based adaptive cluster; CMC = Combined and Multivariate collapsing; skat = SNP-set (Sequence) Kernel Association Method; EDS = Ehlers-Danlos Syndrome.

Cells in bold and with box borders: q-value below 0.05 upon FDR analysis. Hg19 was used as reference genome.

**Supplementary table S4bis:** Results from the gene-based tests, at MAF cutoffs 0.1% , sorted by significance

<b>Gene</b>	<b>p-value</b>	<b>q-value</b>	<b>Test</b>	<b>Variant category</b>	<b>EDS gene</b>
<i>COL2A1</i>	2.00E-04	0.0318	kbac	Intronic variants	non-EDS gene
<i>TNXB</i>	3.00E-04	0.0318	CMC	Nonsynonymous variants and in-frame indels	EDS gene
<i>TNXB</i>	4.00E-04	0.0318	kbac	Nonsynonymous variants and in-frame indels	EDS gene
<i>ZNF469</i>	4.00E-04	0.0318	kbac	Nonsynonymous variants and in-frame indels	EDS gene
<i>TNXB</i>	5.00E-04	0.0318	skat	Nonsynonymous variants and in-frame indels	EDS gene
<i>ZNF469</i>	6.00E-04	0.0318	kbac	Synonymous variants	EDS gene
<i>TNXB</i>	6.00E-04	0.0318	vt	Nonsynonymous variants and in-frame indels	EDS gene
<i>ZNF469</i>	6.00E-04	0.0318	vt	Nonsynonymous variants and in-frame indels	EDS gene
<i>COL5A1</i>	6.00E-04	0.0318	skat	Synonymous variants	EDS gene
<i>COL2A1</i>	0.001	0.0446	vt	Intronic variants	non-EDS gene
<i>COL2A1</i>	0.0013	0.0516	CMC	Intronic variants	non-EDS gene
<i>ZNF469</i>	0.0019	0.0714	CMC	Nonsynonymous variants and in-frame indels	EDS gene
<i>ZNF469</i>	0.003	0.1004	vt	Synonymous variants	EDS gene
<i>ZNF469</i>	0.0032	0.1004	CMC	Synonymous variants	EDS gene
<i>PARP2</i>	0.0043	0.1268	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>MMP9</i>	0.0046	0.1282	kbac	Intronic variants	non-EDS gene
<i>TF</i>	0.0056	0.1365	kbac	Synonymous variants	non-EDS gene
<i>TF</i>	0.0058	0.1365	vt	Synonymous variants	non-EDS gene
<i>COL5A1</i>	0.0058	0.1365	skat	Intronic variants	EDS gene
<i>COL12A1</i>	0.0062	0.1382	vt	Intronic variants	EDS gene
<i>COL12A1</i>	0.0067	0.1396	skat	Synonymous variants	EDS gene
<i>PARP2</i>	0.0072	0.1396	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A1</i>	0.0072	0.1396	kbac	Synonymous variants	EDS gene
<i>COL5A1</i>	0.0078	0.1449	kbac	Intronic variants	EDS gene
<i>MMP9</i>	0.0082	0.1462	calpha	Intronic variants	non-EDS gene
<i>FNDC3B</i>	0.0098	0.1624	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TF</i>	0.0099	0.1624	CMC	Synonymous variants	non-EDS gene
<i>COL5A1</i>	0.0102	0.1624	vt	Intronic variants	EDS gene

<i>COL12A1</i>	0.0106	0.163	kbac	Intronic variants	EDS gene
<i>FNDC3B</i>	0.0117	0.1739	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>ZNF469</i>	0.0124	0.178	skat	Nonsynonymous variants and in-frame indels	EDS gene
<i>DOCK1</i>	0.0149	0.1921	skat	Intronic variants	non-EDS gene
<i>COL12A1</i>	0.0155	0.1921	CMC	Intronic variants	EDS gene
<i>COL12A1</i>	0.0156	0.1921	vt	Synonymous variants	EDS gene
<i>P3H4</i>	0.0156	0.1921	vt	Synonymous variants	non-EDS gene
<i>RDH13</i>	0.0159	0.1921	skat	Intronic variants	non-EDS gene
<i>COL12A1</i>	0.0162	0.1921	kbac	Synonymous variants	EDS gene
<i>COL5A1</i>	0.0168	0.1921	vt	Synonymous variants	EDS gene
<i>COL5A1</i>	0.0168	0.1921	CMC	Intronic variants	EDS gene
<i>FNDC3B</i>	0.0204	0.224	kbac	Intronic variants	non-EDS gene
<i>COL5A1</i>	0.0235	0.224	CMC	Synonymous variants	EDS gene
<i>IL1RN</i>	0.0238	0.224	skat	Synonymous variants	non-EDS gene
<i>FNDC3B</i>	0.024	0.224	kbac	Synonymous variants	non-EDS gene
<i>MMP9</i>	0.024	0.224	vt	Intronic variants	non-EDS gene
<i>IL1RN</i>	0.0244	0.224	vt	Synonymous variants	non-EDS gene
<i>LCA5</i>	0.0253	0.224	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>IL1RN</i>	0.0254	0.224	kbac	Synonymous variants	non-EDS gene
<i>COL5A1</i>	0.026	0.224	calpha	Synonymous variants	EDS gene
<i>VSX1</i>	0.0264	0.224	kbac	Intronic variants	non-EDS gene
<i>P3H4</i>	0.027	0.224	calpha	Synonymous variants	non-EDS gene
<i>VSX1</i>	0.0274	0.224	vt	Intronic variants	non-EDS gene
<i>VSX1</i>	0.0276	0.224	calpha	Intronic variants	non-EDS gene
<i>DOCK1</i>	0.0276	0.224	vt	Intronic variants	non-EDS gene
<i>FNDC3B</i>	0.0278	0.224	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FNDC3B</i>	0.0284	0.224	skat	Intronic variants	non-EDS gene
<i>MMP9</i>	0.0285	0.224	skat	Intronic variants	non-EDS gene
<i>ZEB1</i>	0.0286	0.224	skat	Synonymous variants	non-EDS gene
<i>PARP2</i>	0.0294	0.226	vt	Nonsynonymous variants and in-frame indels	non-EDS gene

<i>ZEB1</i>	0.031	0.2318	kbac	Intronic variants	non-EDS gene
<i>MMP9</i>	0.0312	0.2318	CMC	Intronic variants	non-EDS gene
<i>SOD1</i>	0.0334	0.2392	calpha	5' UTR variants	non-EDS gene
<i>RDH13</i>	0.0334	0.2392	kbac	Intronic variants	non-EDS gene
<i>LCA5</i>	0.0338	0.2392	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP2</i>	0.035	0.2441	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL2A1</i>	0.0368	0.2486	calpha	Intronic variants	non-EDS gene
<i>ZNF469</i>	0.0368	0.2486	calpha	Synonymous variants	EDS gene
<i>LCA5</i>	0.0382	0.2494	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TF</i>	0.0382	0.2494	skat	Synonymous variants	non-EDS gene
<i>MMP9</i>	0.0386	0.2494	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>P3H4</i>	0.0396	0.2522	kbac	Synonymous variants	non-EDS gene
<i>SLC4A11</i>	0.0426	0.2606	kbac	Synonymous variants	non-EDS gene
<i>TNXB</i>	0.0435	0.2606	skat	Synonymous variants	EDS gene
<i>PARP2</i>	0.0442	0.2606	vt	Synonymous variants	non-EDS gene
<i>RDH13</i>	0.0446	0.2606	skat	Synonymous variants	non-EDS gene
<i>FNDC3B</i>	0.0449	0.2606	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TGFBI</i>	0.0452	0.2606	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL12A1</i>	0.0455	0.2606	CMC	Synonymous variants	EDS gene
<i>RDH13</i>	0.0468	0.2606	calpha	Synonymous variants	non-EDS gene
<i>ZNF469</i>	0.047	0.2606	calpha	Nonsynonymous variants and in-frame indels	EDS gene
<i>SOD1</i>	0.0476	0.2606	skat	Intronic variants	non-EDS gene
<i>ZEB1</i>	0.0484	0.2606	kbac	Synonymous variants	non-EDS gene
<i>DOCK1</i>	0.0486	0.2606	vt	Synonymous variants	non-EDS gene
<i>ZEB1</i>	0.05	0.2606	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>ZEB1</i>	0.0502	0.2606	vt	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.0508	0.2606	calpha	Synonymous variants	non-EDS gene
<i>PARP2</i>	0.0514	0.2606	kbac	Synonymous variants	non-EDS gene
<i>MMP9</i>	0.052	0.2606	skat	Synonymous variants	non-EDS gene
<i>VSX1</i>	0.0523	0.2606	CMC	Intronic variants	non-EDS gene

<i>IL1RN</i>	0.0525	0.2606	CMC	Synonymous variants	non-EDS gene
<i>TF</i>	0.0526	0.2606	calpha	Synonymous variants	non-EDS gene
<i>IL1RN</i>	0.054	0.2646	calpha	Synonymous variants	non-EDS gene
<i>LOX</i>	0.0548	0.2656	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.0556	0.2665	kbac	Synonymous variants	non-EDS gene
<i>IL1RN</i>	0.0582	0.2713	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL12A1</i>	0.0586	0.2713	vt	Nonsynonymous variants and in-frame indels	EDS gene
<i>FNDC3B</i>	0.0592	0.2713	vt	Intronic variants	non-EDS gene
<i>P3H4</i>	0.0598	0.2713	CMC	Synonymous variants	non-EDS gene
<i>COL12A1</i>	0.06	0.2713	kbac	Nonsynonymous variants and in-frame indels	EDS gene
<i>VSX1</i>	0.0606	0.2713	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.0614	0.2713	kbac	Synonymous variants	non-EDS gene
<i>SOD1</i>	0.0624	0.2713	calpha	Intronic variants	non-EDS gene
<i>TGFBI</i>	0.0628	0.2713	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.0632	0.2713	calpha	Nonsynonymous variants and in-frame indels	EDS gene
<i>NLRP1</i>	0.0633	0.2713	skat	Synonymous variants	non-EDS gene
<i>DOCK1</i>	0.0654	0.2726	kbac	Synonymous variants	non-EDS gene
<i>MMP9</i>	0.0654	0.2726	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>ZEB1</i>	0.0658	0.2726	calpha	Synonymous variants	non-EDS gene
<i>TNXB</i>	0.068	0.2726	calpha	Intronic variants	EDS gene
<i>TF</i>	0.0684	0.2726	kbac	Intronic variants	non-EDS gene
<i>SLC4A11</i>	0.0688	0.2726	skat	5' UTR variants	non-EDS gene
<i>ZEB1</i>	0.0692	0.2726	calpha	Intronic variants	non-EDS gene
<i>DOCK1</i>	0.07	0.2726	kbac	Intronic variants	non-EDS gene
<i>FNDC3B</i>	0.0701	0.2726	CMC	Intronic variants	non-EDS gene
<i>DOCK1</i>	0.0701	0.2726	CMC	Synonymous variants	non-EDS gene
<i>DOCK1</i>	0.0714	0.2726	CMC	Intronic variants	non-EDS gene
<i>TF</i>	0.0714	0.2726	vt	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.0715	0.2726	CMC	Synonymous variants	non-EDS gene
<i>LOX</i>	0.0738	0.2788	vt	Nonsynonymous variants and in-frame indels	non-EDS gene

<i>LOXHD1</i>	0.0746	0.2793	vt	Synonymous variants	non-EDS gene
<i>FNDC3B</i>	0.0756	0.2793	vt	Synonymous variants	non-EDS gene
<i>COL5A1</i>	0.0758	0.2793	calpha	Intronic variants	EDS gene
<i>VSX1</i>	0.077	0.2814	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>EPPK1</i>	0.081	0.2936	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK9</i>	0.0828	0.2977	vt	Intronic variants	non-EDS gene
<i>LAMC3</i>	0.0846	0.3017	kbac	Intronic variants	non-EDS gene
<i>COL12A1</i>	0.0863	0.3044	CMC	Nonsynonymous variants and in-frame indels	EDS gene
<i>ZEB1</i>	0.0868	0.3044	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.0874	0.3044	vt	Synonymous variants	non-EDS gene
<i>PARP2</i>	0.0888	0.3045	calpha	Synonymous variants	non-EDS gene
<i>IL1RN</i>	0.0888	0.3045	vt	Intronic variants	non-EDS gene
<i>MMP9</i>	0.0908	0.307	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LAMC3</i>	0.0916	0.307	kbac	Synonymous variants	non-EDS gene
<i>IL1RN</i>	0.0922	0.307	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FNDC3B</i>	0.0931	0.307	skat	Synonymous variants	non-EDS gene
<i>TF</i>	0.0943	0.307	skat	Intronic variants	non-EDS gene
<i>MMP9</i>	0.0954	0.307	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>EPPK1</i>	0.0962	0.307	kbac	Synonymous variants	non-EDS gene
<i>FNDC3B</i>	0.0962	0.307	CMC	Synonymous variants	non-EDS gene
<i>PARP2</i>	0.0962	0.307	CMC	Synonymous variants	non-EDS gene
<i>RDH13</i>	0.0964	0.307	calpha	Intronic variants	non-EDS gene
<i>WNT10A</i>	0.0971	0.3072	skat	5' UTR variants	non-EDS gene
<i>SLC4A11</i>	0.0988	0.3082	vt	5' UTR variants	non-EDS gene
<i>VSX1</i>	0.0988	0.3082	skat	Intronic variants	non-EDS gene
<i>SLC4A11</i>	0.103	0.3124	kbac	5' UTR variants	non-EDS gene
<i>IL1RN</i>	0.1038	0.3124	kbac	Intronic variants	non-EDS gene
<i>COL5A3</i>	0.104	0.3124	vt	Synonymous variants	non-EDS gene
<i>LOXHD1</i>	0.104	0.3124	CMC	Synonymous variants	non-EDS gene
<i>RDH13</i>	0.1044	0.3124	vt	Nonsense and frameshift variants	non-EDS gene



<i>LCA5</i>	0.1058	0.3124	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TGFBI</i>	0.1058	0.3124	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.1058	0.3124	calpha	5' UTR variants	non-EDS gene
<i>ZEB1</i>	0.1104	0.3185	CMC	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.1112	0.3185	kbac	Synonymous variants	non-EDS gene
<i>TNXB</i>	0.1113	0.3185	CMC	Intronic variants	EDS gene
<i>LOXHD1</i>	0.1146	0.3185	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.1148	0.3185	skat	5' UTR variants	non-EDS gene
<i>IL1RN</i>	0.115	0.3185	skat	Intronic variants	non-EDS gene
<i>PARP1</i>	0.1153	0.3185	calpha	Synonymous variants	non-EDS gene
<i>RDH13</i>	0.1179	0.3185	calpha	Nonsense and frameshift variants	non-EDS gene
<i>FOXO1</i>	0.118	0.3185	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>HLA-G</i>	0.1191	0.3185	skat	Intronic variants	non-EDS gene
<i>IL1RN</i>	0.1209	0.3185	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>HLA-G</i>	0.1209	0.3185	kbac	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.1209	0.3185	kbac	Intronic variants	non-EDS gene
<i>COL5A3</i>	0.1214	0.3185	skat	3' UTR variants	non-EDS gene
<i>EPPK1</i>	0.1219	0.3185	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>EPPK1</i>	0.1219	0.3185	vt	Synonymous variants	non-EDS gene
<i>PARP2</i>	0.1234	0.3185	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.1239	0.3185	vt	Intronic variants	non-EDS gene
<i>NLRP1</i>	0.1241	0.3185	skat	3' UTR variants	non-EDS gene
<i>LOX</i>	0.1249	0.3185	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.1249	0.3185	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL12A1</i>	0.1252	0.3185	skat	Intronic variants	EDS gene
<i>LAMC3</i>	0.1259	0.3185	vt	Synonymous variants	non-EDS gene
<i>MIR184</i>	0.1268	0.3185	skat	noncoding RNA 5'	non-EDS gene
<i>NLRP1</i>	0.1269	0.3185	kbac	3' UTR variants	non-EDS gene
<i>HLA-G</i>	0.127	0.3185	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.1279	0.3185	vt	Intronic variants	non-EDS gene

<i>DOCK1</i>	0.1289	0.3185	calpha	Intronic variants	non-EDS gene
<i>RDH13</i>	0.1289	0.3185	kbac	Nonsense and frameshift variants	non-EDS gene
<i>RDH13</i>	0.1301	0.3185	CMC	Intronic variants	non-EDS gene
<i>SLC4A11</i>	0.1304	0.3185	CMC	Synonymous variants	non-EDS gene
<i>LAMC3</i>	0.1309	0.3185	vt	Intronic variants	non-EDS gene
<i>ZEB1</i>	0.1324	0.3185	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TGFBI</i>	0.1329	0.3185	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.1329	0.3185	kbac	Intronic variants	non-EDS gene
<i>MMP9</i>	0.1374	0.3258	kbac	Synonymous variants	non-EDS gene
<i>ZEB1</i>	0.1379	0.3258	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.1389	0.3258	calpha	Synonymous variants	non-EDS gene
<i>DCAF11</i>	0.1389	0.3258	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>EPPK1</i>	0.1395	0.3258	skat	Nonsense and frameshift variants	non-EDS gene
<i>LAMC3</i>	0.1419	0.328	skat	Synonymous variants	non-EDS gene
<i>FOXO1</i>	0.1423	0.328	skat	Synonymous variants	non-EDS gene
<i>ZEB1</i>	0.1427	0.328	skat	Intronic variants	non-EDS gene
<i>LAMC3</i>	0.1459	0.3318	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.1459	0.3318	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.1498	0.3391	skat	5' UTR variants	non-EDS gene
<i>NLRP1</i>	0.1528	0.343	calpha	3' UTR variants	non-EDS gene
<i>COL5A3</i>	0.1533	0.343	CMC	Synonymous variants	non-EDS gene
<i>SOD1</i>	0.1552	0.343	skat	5' UTR variants	non-EDS gene
<i>WNT10A</i>	0.1556	0.343	skat	Synonymous variants	non-EDS gene
<i>ZEB1</i>	0.1564	0.343	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>P3H4</i>	0.1578	0.343	kbac	Intronic variants	non-EDS gene
<i>TNXB</i>	0.1585	0.343	skat	Nonsense and frameshift variants	EDS gene
<i>LAMC3</i>	0.1598	0.343	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>WNT10A</i>	0.16	0.343	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FNDC3B</i>	0.1628	0.343	calpha	Synonymous variants	non-EDS gene
<i>IL1RN</i>	0.1631	0.343	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene

<i>SLC4A11</i>	0.1634	0.343	skat	Synonymous variants	non-EDS gene
<i>LOX</i>	0.1636	0.343	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.1638	0.343	calpha	Nonsense and frameshift variants	non-EDS gene
<i>COL12A1</i>	0.1638	0.343	calpha	Intronic variants	EDS gene
<i>VSX1</i>	0.1638	0.343	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>P3H4</i>	0.1668	0.3476	vt	Intronic variants	non-EDS gene
<i>LAMC3</i>	0.1714	0.3555	CMC	Intronic variants	non-EDS gene
<i>FOXO1</i>	0.1738	0.3588	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.1808	0.3657	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DCAF11</i>	0.1818	0.3657	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>MMP9</i>	0.1838	0.3657	vt	Synonymous variants	non-EDS gene
<i>ZEB1</i>	0.1838	0.3657	vt	Synonymous variants	non-EDS gene
<i>ZEB1</i>	0.1842	0.3657	skat	Nonsense and frameshift variants	non-EDS gene
<i>COL5A3</i>	0.1848	0.3657	kbac	Intronic variants	non-EDS gene
<i>VSX1</i>	0.1853	0.3657	skat	Nonsense and frameshift variants	non-EDS gene
<i>LAMC3</i>	0.1858	0.3657	CMC	Synonymous variants	non-EDS gene
<i>COL12A1</i>	0.1863	0.3657	skat	Nonsynonymous variants and in-frame indels	EDS gene
<i>COL12A1</i>	0.1864	0.3657	skat	Nonsense and frameshift variants	EDS gene
<i>FASLG</i>	0.1876	0.3657	skat	Synonymous variants	non-EDS gene
<i>FNDC3B</i>	0.1878	0.3657	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK9</i>	0.1878	0.3657	vt	Synonymous variants	non-EDS gene
<i>DOCK9</i>	0.1894	0.3672	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LCA5</i>	0.1938	0.3735	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>ZEB1</i>	0.1945	0.3735	CMC	Synonymous variants	non-EDS gene
<i>DCAF11</i>	0.1958	0.3735	kbac	Synonymous variants	non-EDS gene
<i>LOXHD1</i>	0.1968	0.3735	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK9</i>	0.1971	0.3735	CMC	Intronic variants	non-EDS gene
<i>RDH13</i>	0.1977	0.3735	skat	Nonsense and frameshift variants	non-EDS gene
<i>EPPK1</i>	0.1986	0.3735	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.1998	0.3735	calpha	Synonymous variants	non-EDS gene

<i>VSX1</i>	0.2002	0.3735	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.2028	0.3755	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.2029	0.3755	CMC	5' UTR variants	non-EDS gene
<i>IL1RN</i>	0.2068	0.381	calpha	Intronic variants	non-EDS gene
<i>NLRP1</i>	0.2094	0.3842	skat	Nonsense and frameshift variants	non-EDS gene
<i>DCAF11</i>	0.222	0.4046	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TF</i>	0.2223	0.4046	CMC	Intronic variants	non-EDS gene
<i>LAMC3</i>	0.2261	0.4094	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.2268	0.4094	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.2294	0.4118	CMC	Nonsense and frameshift variants	non-EDS gene
<i>COL5A3</i>	0.23	0.4118	CMC	3' UTR variants	non-EDS gene
<i>MIR184</i>	0.233	0.4141	CMC	noncoding RNA 5'	non-EDS gene
<i>HLA-G</i>	0.2331	0.4141	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>WNT10A</i>	0.2358	0.4172	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.2399	0.4227	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.2418	0.4244	kbac	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.2448	0.4282	skat	Intronic variants	non-EDS gene
<i>VSX1</i>	0.2465	0.4293	skat	3' UTR variants	non-EDS gene
<i>FOXO1</i>	0.2507	0.4335	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.2508	0.4335	CMC	Nonsense and frameshift variants	non-EDS gene
<i>TNXB</i>	0.2537	0.4369	calpha	Synonymous variants	EDS gene
<i>MMP9</i>	0.2567	0.4403	calpha	Synonymous variants	non-EDS gene
<i>DOCK1</i>	0.2585	0.4416	skat	Synonymous variants	non-EDS gene
<i>SLC4A11</i>	0.2594	0.4416	CMC	Intronic variants	non-EDS gene
<i>LOXHD1</i>	0.2687	0.4532	kbac	Intronic variants	non-EDS gene
<i>FOXO1</i>	0.2701	0.4532	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>IL1RN</i>	0.2701	0.4532	CMC	Intronic variants	non-EDS gene
<i>EPPK1</i>	0.2712	0.4532	CMC	Synonymous variants	non-EDS gene
<i>DOCK9</i>	0.2714	0.4532	CMC	Synonymous variants	non-EDS gene
<i>PARP1</i>	0.2769	0.4595	skat	Intronic variants	non-EDS gene

<i>FOXO1</i>	0.2777	0.4595	calpha	Synonymous variants	non-EDS gene
<i>COL5A3</i>	0.2782	0.4595	skat	Intronic variants	non-EDS gene
<i>COL5A1</i>	0.2832	0.466	skat	Nonsynonymous variants and in-frame indels	EDS gene
<i>DOCK1</i>	0.2887	0.4714	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.2897	0.4714	vt	Nonsense and frameshift variants	non-EDS gene
<i>DOCK9</i>	0.2907	0.4714	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>EPPK1</i>	0.2907	0.4714	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.293	0.4734	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>VSX1</i>	0.2967	0.4759	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL2A1</i>	0.2967	0.4759	vt	Synonymous variants	non-EDS gene
<i>TNXB</i>	0.2987	0.4774	kbac	Synonymous variants	EDS gene
<i>PARP2</i>	0.3054	0.4851	skat	Intronic variants	non-EDS gene
<i>COL5A3</i>	0.3057	0.4851	vt	Intronic variants	non-EDS gene
<i>TF</i>	0.3107	0.4913	calpha	Intronic variants	non-EDS gene
<i>DCAF11</i>	0.3159	0.4978	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.3183	0.4998	skat	Nonsense and frameshift variants	non-EDS gene
<i>LOXHD1</i>	0.3197	0.5002	kbac	Nonsense and frameshift variants	non-EDS gene
<i>RDH13</i>	0.3237	0.5047	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP2</i>	0.3267	0.5058	kbac	Intronic variants	non-EDS gene
<i>HLA-G</i>	0.3267	0.5058	vt	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.3357	0.5179	calpha	3' UTR variants	non-EDS gene
<i>COL12A1</i>	0.3397	0.5223	calpha	Synonymous variants	EDS gene
<i>COL5A3</i>	0.3437	0.526	calpha	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.3457	0.526	calpha	3' UTR variants	non-EDS gene
<i>SOD1</i>	0.3457	0.526	vt	Intronic variants	non-EDS gene
<i>LOX</i>	0.3516	0.5293	calpha	3' UTR variants	non-EDS gene
<i>DCAF11</i>	0.3516	0.5293	kbac	3' UTR variants	non-EDS gene
<i>TNXB</i>	0.3536	0.5293	vt	Synonymous variants	EDS gene
<i>P3H4</i>	0.3541	0.5293	CMC	Intronic variants	non-EDS gene
<i>DCAF11</i>	0.3546	0.5293	calpha	Intronic variants	non-EDS gene

<i>MMP9</i>	0.3549	0.5293	CMC	Synonymous variants	non-EDS gene
<i>PRPH2</i>	0.3626	0.538	calpha	Intronic variants	non-EDS gene
<i>DOCK1</i>	0.3666	0.538	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.3676	0.538	vt	Intronic variants	non-EDS gene
<i>WNT10A</i>	0.3685	0.538	CMC	5' UTR variants	non-EDS gene
<i>NLRP1</i>	0.3696	0.538	vt	Intronic variants	non-EDS gene
<i>SLC4A11</i>	0.3706	0.538	calpha	Intronic variants	non-EDS gene
<i>DCAF11</i>	0.3706	0.538	vt	Synonymous variants	non-EDS gene
<i>COL5A3</i>	0.3715	0.538	skat	Synonymous variants	non-EDS gene
<i>DOCK1</i>	0.3726	0.538	calpha	Synonymous variants	non-EDS gene
<i>COL5A3</i>	0.3739	0.538	CMC	Intronic variants	non-EDS gene
<i>NLRP1</i>	0.3756	0.538	kbac	Synonymous variants	non-EDS gene
<i>COL2A1</i>	0.3766	0.538	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK9</i>	0.3774	0.538	skat	Synonymous variants	non-EDS gene
<i>LOXHD1</i>	0.3776	0.538	vt	Intronic variants	non-EDS gene
<i>LAMC3</i>	0.3837	0.5449	skat	Intronic variants	non-EDS gene
<i>P3H4</i>	0.3866	0.5473	calpha	Intronic variants	non-EDS gene
<i>COL2A1</i>	0.3966	0.558	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.3967	0.558	skat	Synonymous variants	non-EDS gene
<i>TF</i>	0.4036	0.5634	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.4036	0.5634	CMC	5' UTR variants	non-EDS gene
<i>EPPK1</i>	0.4044	0.5634	skat	Synonymous variants	non-EDS gene
<i>FNDC3B</i>	0.4056	0.5634	calpha	Intronic variants	non-EDS gene
<i>RDH13</i>	0.4096	0.5672	kbac	Synonymous variants	non-EDS gene
<i>DCAF11</i>	0.4146	0.5723	vt	3' UTR variants	non-EDS gene
<i>LOXHD1</i>	0.4206	0.5771	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LAMC3</i>	0.4206	0.5771	calpha	Synonymous variants	non-EDS gene
<i>COL5A1</i>	0.4246	0.5804	kbac	Nonsynonymous variants and in-frame indels	EDS gene
<i>FOXO1</i>	0.4276	0.5804	kbac	Synonymous variants	non-EDS gene
<i>P3H4</i>	0.43	0.5804	skat	Intronic variants	non-EDS gene

<i>NLRP1</i>	0.4326	0.5804	kbac	Intronic variants	non-EDS gene
<i>COL5A3</i>	0.4343	0.5804	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.4348	0.5804	CMC	3' UTR variants	non-EDS gene
<i>HLA-G</i>	0.4348	0.5804	CMC	Intronic variants	non-EDS gene
<i>DCAF11</i>	0.4352	0.5804	skat	Synonymous variants	non-EDS gene
<i>SLC4A11</i>	0.4368	0.5804	skat	Intronic variants	non-EDS gene
<i>TGFBI</i>	0.439	0.5804	skat	Intronic variants	non-EDS gene
<i>EPPK1</i>	0.4401	0.5804	CMC	Nonsense and frameshift variants	non-EDS gene
<i>COL2A1</i>	0.4416	0.5804	kbac	Synonymous variants	non-EDS gene
<i>LAMC3</i>	0.4426	0.5804	calpha	Intronic variants	non-EDS gene
<i>DOCK1</i>	0.4436	0.5804	kbac	3' UTR variants	non-EDS gene
<i>TGFBI</i>	0.4436	0.5804	vt	Intronic variants	non-EDS gene
<i>EPPK1</i>	0.4496	0.5804	calpha	Synonymous variants	non-EDS gene
<i>P3H4</i>	0.4496	0.5804	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.4525	0.5804	vt	Synonymous variants	non-EDS gene
<i>TGFBI</i>	0.4535	0.5804	kbac	Intronic variants	non-EDS gene
<i>PARP2</i>	0.4545	0.5804	vt	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.4549	0.5804	CMC	3' UTR variants	non-EDS gene
<i>TGFBI</i>	0.4575	0.5804	kbac	Synonymous variants	non-EDS gene
<i>DOCK1</i>	0.4575	0.5804	vt	3' UTR variants	non-EDS gene
<i>PARP1</i>	0.458	0.5804	CMC	Intronic variants	non-EDS gene
<i>RDH13</i>	0.4583	0.5804	CMC	5' UTR variants	non-EDS gene
<i>VSX1</i>	0.4585	0.5804	kbac	Synonymous variants	non-EDS gene
<i>TF</i>	0.4586	0.5804	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FASLG</i>	0.4595	0.5804	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>WNT10A</i>	0.4608	0.5804	CMC	Synonymous variants	non-EDS gene
<i>TNXB</i>	0.4651	0.5842	CMC	Nonsense and frameshift variants	EDS gene
<i>DOCK1</i>	0.4709	0.5858	skat	3' UTR variants	non-EDS gene
<i>SOD1</i>	0.4725	0.5858	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>WNT10A</i>	0.4725	0.5858	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene

<i>LCA5</i>	0.4765	0.5858	vt	Synonymous variants	non-EDS gene
<i>ZEB1</i>	0.4791	0.5858	CMC	Nonsense and frameshift variants	non-EDS gene
<i>VSX1</i>	0.4794	0.5858	CMC	Nonsense and frameshift variants	non-EDS gene
<i>TGFBI</i>	0.4795	0.5858	vt	Synonymous variants	non-EDS gene
<i>LOXHD1</i>	0.4796	0.5858	CMC	Intronic variants	non-EDS gene
<i>COL12A1</i>	0.4797	0.5858	CMC	Nonsense and frameshift variants	EDS gene
<i>FASLG</i>	0.48	0.5858	CMC	Synonymous variants	non-EDS gene
<i>COL5A3</i>	0.4835	0.5858	calpha	Synonymous variants	non-EDS gene
<i>NLRP1</i>	0.4844	0.5858	CMC	Nonsense and frameshift variants	non-EDS gene
<i>FOXO1</i>	0.4845	0.5858	vt	Synonymous variants	non-EDS gene
<i>RDH13</i>	0.4855	0.5858	vt	Synonymous variants	non-EDS gene
<i>FASLG</i>	0.4885	0.5858	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.4885	0.5858	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>VSX1</i>	0.4887	0.5858	CMC	3' UTR variants	non-EDS gene
<i>DCAF11</i>	0.4912	0.5872	CMC	Synonymous variants	non-EDS gene
<i>SOD1</i>	0.4935	0.5884	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PRPH2</i>	0.4997	0.592	CMC	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.5003	0.592	CMC	3' UTR variants	non-EDS gene
<i>LOX</i>	0.5006	0.592	CMC	3' UTR variants	non-EDS gene
<i>LOX</i>	0.5075	0.5987	calpha	Synonymous variants	non-EDS gene
<i>DOCK9</i>	0.5095	0.5994	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.5115	0.6002	calpha	5' UTR variants	non-EDS gene
<i>LOXHD1</i>	0.5135	0.601	calpha	Nonsense and frameshift variants	non-EDS gene
<i>LOXHD1</i>	0.5155	0.6017	calpha	Synonymous variants	non-EDS gene
<i>VSX1</i>	0.5185	0.6036	vt	Synonymous variants	non-EDS gene
<i>NLRP1</i>	0.5205	0.6044	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.5271	0.6105	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.5335	0.6147	calpha	Intronic variants	non-EDS gene
<i>NLRP1</i>	0.5335	0.6147	vt	3' UTR variants	non-EDS gene
<i>COL5A1</i>	0.5455	0.6269	vt	Nonsynonymous variants and in-frame indels	EDS gene



<i>VSX1</i>	0.5504	0.6307	calpha	Synonymous variants	non-EDS gene
<i>HLA-G</i>	0.5524	0.6307	calpha	Intronic variants	non-EDS gene
<i>FASLG</i>	0.5536	0.6307	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>VSX1</i>	0.5556	0.6307	skat	Synonymous variants	non-EDS gene
<i>LCA5</i>	0.5559	0.6307	skat	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.5684	0.6433	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP2</i>	0.5714	0.6451	calpha	Intronic variants	non-EDS gene
<i>LOX</i>	0.5755	0.648	skat	Nonsense and frameshift variants	non-EDS gene
<i>RDH13</i>	0.5838	0.6541	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.5838	0.6541	CMC	Synonymous variants	non-EDS gene
<i>DOCK9</i>	0.5914	0.6609	calpha	Synonymous variants	non-EDS gene
<i>P3H4</i>	0.5964	0.6638	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LCA5</i>	0.5974	0.6638	vt	Intronic variants	non-EDS gene
<i>SOD1</i>	0.5984	0.6638	kbac	Intronic variants	non-EDS gene
<i>WNT10A</i>	0.6004	0.6643	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TGFBI</i>	0.6047	0.6675	skat	Synonymous variants	non-EDS gene
<i>LCA5</i>	0.6074	0.6677	kbac	Intronic variants	non-EDS gene
<i>DOCK1</i>	0.6079	0.6677	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.6094	0.6677	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.6154	0.6726	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PRPH2</i>	0.6214	0.6747	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FASLG</i>	0.6229	0.6747	skat	5' UTR variants	non-EDS gene
<i>LOX</i>	0.6257	0.6747	CMC	5' UTR variants	non-EDS gene
<i>PRPH2</i>	0.6258	0.6747	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.6258	0.6747	CMC	Synonymous variants	non-EDS gene
<i>DCAF11</i>	0.6264	0.6747	kbac	Intronic variants	non-EDS gene
<i>COL2A1</i>	0.6313	0.6761	skat	Intronic variants	non-EDS gene
<i>COL5A3</i>	0.6316	0.6761	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LCA5</i>	0.6334	0.6761	kbac	Synonymous variants	non-EDS gene
<i>LOXHD1</i>	0.636	0.6761	skat	Nonsynonymous variants and in-frame indels	non-EDS gene

<i>TF</i>	0.6361	0.6761	skat	5' UTR variants	non-EDS gene
<i>PARP2</i>	0.6368	0.6761	CMC	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.643	0.6793	skat	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.6434	0.6793	calpha	Intronic variants	non-EDS gene
<i>SLC4A11</i>	0.6444	0.6793	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A1</i>	0.6464	0.6798	calpha	Nonsynonymous variants and in-frame indels	EDS gene
<i>PARP2</i>	0.6529	0.685	skat	Synonymous variants	non-EDS gene
<i>TGFBI</i>	0.6593	0.6897	calpha	Intronic variants	non-EDS gene
<i>NLRP1</i>	0.6604	0.6897	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.6628	0.6905	skat	Intronic variants	non-EDS gene
<i>DCAF11</i>	0.6693	0.6957	vt	Intronic variants	non-EDS gene
<i>LOXHD1</i>	0.6751	0.6986	CMC	Nonsense and frameshift variants	non-EDS gene
<i>DCAF11</i>	0.6753	0.6986	CMC	3' UTR variants	non-EDS gene
<i>LOX</i>	0.6773	0.6991	kbac	Intronic variants	non-EDS gene
<i>EPPK1</i>	0.682	0.7023	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.6873	0.7046	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.6873	0.7046	vt	Intronic variants	non-EDS gene
<i>SOD1</i>	0.6897	0.7048	CMC	5' UTR variants	non-EDS gene
<i>DCAF11</i>	0.6907	0.7048	skat	3' UTR variants	non-EDS gene
<i>COL5A1</i>	0.702	0.7147	CMC	Nonsynonymous variants and in-frame indels	EDS gene
<i>SOD1</i>	0.7092	0.7203	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DCAF11</i>	0.7203	0.7293	calpha	3' UTR variants	non-EDS gene
<i>LAMC3</i>	0.7213	0.7293	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DCAF11</i>	0.7233	0.7297	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FASLG</i>	0.7293	0.7341	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DCAF11</i>	0.7343	0.7374	calpha	Synonymous variants	non-EDS gene
<i>LOX</i>	0.7398	0.7388	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SOD1</i>	0.7433	0.7388	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SOD1</i>	0.7433	0.7388	kbac	5' UTR variants	non-EDS gene
<i>TNXB</i>	0.7447	0.7388	CMC	Synonymous variants	EDS gene

<i>LOXHD1</i>	0.7452	0.7388	skat	Intronic variants	non-EDS gene
<i>LCA5</i>	0.7463	0.7388	calpha	Synonymous variants	non-EDS gene
<i>COL12A1</i>	0.7473	0.7388	calpha	Nonsynonymous variants and in-frame indels	EDS gene
<i>LOX</i>	0.7542	0.7441	vt	Synonymous variants	non-EDS gene
<i>LOX</i>	0.7582	0.7464	kbac	5' UTR variants	non-EDS gene
<i>COL2A1</i>	0.7662	0.7503	CMC	Synonymous variants	non-EDS gene
<i>P3H4</i>	0.7686	0.7503	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LCA5</i>	0.7707	0.7503	skat	Synonymous variants	non-EDS gene
<i>DCAF11</i>	0.7707	0.7503	skat	Intronic variants	non-EDS gene
<i>PARP1</i>	0.7742	0.7503	kbac	Synonymous variants	non-EDS gene
<i>LOX</i>	0.7749	0.7503	skat	5' UTR variants	non-EDS gene
<i>P3H4</i>	0.7765	0.7503	skat	Synonymous variants	non-EDS gene
<i>COL2A1</i>	0.7772	0.7503	calpha	Synonymous variants	non-EDS gene
<i>DOCK9</i>	0.7791	0.7503	skat	5' UTR variants	non-EDS gene
<i>PARP1</i>	0.7792	0.7503	vt	Synonymous variants	non-EDS gene
<i>LOX</i>	0.7892	0.7503	vt	5' UTR variants	non-EDS gene
<i>LOX</i>	0.7902	0.7503	kbac	Synonymous variants	non-EDS gene
<i>FNDC3B</i>	0.7922	0.7503	skat	3' UTR variants	non-EDS gene
<i>SOD1</i>	0.7922	0.7503	skat	Synonymous variants	non-EDS gene
<i>IL1RN</i>	0.7924	0.7503	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>HLA-G</i>	0.7939	0.7503	skat	Synonymous variants	non-EDS gene
<i>TF</i>	0.7942	0.7503	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>MMP9</i>	0.7965	0.7503	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>VSX1</i>	0.7969	0.7503	CMC	Synonymous variants	non-EDS gene
<i>COL2A1</i>	0.7969	0.7503	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LCA5</i>	0.7984	0.7503	skat	3' UTR variants	non-EDS gene
<i>DOCK1</i>	0.7992	0.7503	calpha	3' UTR variants	non-EDS gene
<i>TGFBI</i>	0.8024	0.7517	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.8084	0.7557	CMC	Synonymous variants	non-EDS gene
<i>SOD1</i>	0.8102	0.7558	vt	5' UTR variants	non-EDS gene

<i>COL2A1</i>	0.815	0.7587	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.8182	0.7588	CMC	Intronic variants	non-EDS gene
<i>FOXO1</i>	0.8185	0.7588	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL2A1</i>	0.8227	0.7611	skat	Synonymous variants	non-EDS gene
<i>TF</i>	0.8372	0.765	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.8379	0.765	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.839	0.765	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL2A1</i>	0.8432	0.765	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PRPH2</i>	0.8482	0.765	kbac	3' UTR variants	non-EDS gene
<i>PRPH2</i>	0.8511	0.765	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>P3H4</i>	0.8521	0.765	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK9</i>	0.8531	0.765	vt	3' UTR variants	non-EDS gene
<i>LOX</i>	0.8561	0.765	kbac	3' UTR variants	non-EDS gene
<i>PRPH2</i>	0.8568	0.765	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TGFBI</i>	0.8571	0.765	calpha	Synonymous variants	non-EDS gene
<i>LCA5</i>	0.8591	0.765	calpha	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.8591	0.765	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PRPH2</i>	0.8619	0.765	skat	3' UTR variants	non-EDS gene
<i>DOCK9</i>	0.8623	0.765	skat	3' UTR variants	non-EDS gene
<i>NLRP1</i>	0.8629	0.765	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.8631	0.765	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.8631	0.765	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.8647	0.765	skat	Intronic variants	EDS gene
<i>LOX</i>	0.8661	0.765	vt	3' UTR variants	non-EDS gene
<i>PARP1</i>	0.8664	0.765	skat	Synonymous variants	non-EDS gene
<i>PRPH2</i>	0.8671	0.765	kbac	Intronic variants	non-EDS gene
<i>TF</i>	0.8674	0.765	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PRPH2</i>	0.8681	0.765	vt	3' UTR variants	non-EDS gene
<i>ZNF469</i>	0.872	0.7664	skat	Synonymous variants	EDS gene
<i>DOCK9</i>	0.8731	0.7664	kbac	3' UTR variants	non-EDS gene

<i>LOX</i>	0.8754	0.7669	skat	3' UTR variants	non-EDS gene
<i>PRPH2</i>	0.8851	0.7732	vt	Intronic variants	non-EDS gene
<i>RDH13</i>	0.8861	0.7732	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.8892	0.7744	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.8925	0.7748	skat	Intronic variants	non-EDS gene
<i>LOX</i>	0.8931	0.7748	skat	Synonymous variants	non-EDS gene
<i>PARP1</i>	0.9	0.7787	skat	Nonsense and frameshift variants	non-EDS gene
<i>PRPH2</i>	0.901	0.7787	skat	Synonymous variants	non-EDS gene
<i>SLC4A11</i>	0.9111	0.7853	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LAMC3</i>	0.9123	0.7853	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.9171	0.7879	calpha	Intronic variants	non-EDS gene
<i>PARP1</i>	0.9231	0.7916	vt	Nonsense and frameshift variants	non-EDS gene
<i>NLRP1</i>	0.9311	0.7969	calpha	Intronic variants	non-EDS gene
<i>LOXHD1</i>	0.9421	0.8047	calpha	Intronic variants	non-EDS gene
<i>PARP1</i>	0.9481	0.8083	kbac	Nonsense and frameshift variants	non-EDS gene
<i>PRPH2</i>	0.964	0.8093	kbac	Synonymous variants	non-EDS gene
<i>TNXB</i>	0.973	0.8093	kbac	Intronic variants	EDS gene
<i>TNXB</i>	0.977	0.8093	vt	Intronic variants	EDS gene
<i>PRPH2</i>	0.993	0.8093	vt	Synonymous variants	non-EDS gene
<i>DOCK9</i>	1	0.8093	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FASLG</i>	1	0.8093	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>P3H4</i>	1	0.8093	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SOD1</i>	1	0.8093	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	1	0.8093	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>WNT10A</i>	1	0.8093	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	1	0.8093	CMC	Nonsense and frameshift variants	non-EDS gene
<i>DOCK1</i>	1	0.8093	CMC	3' UTR variants	non-EDS gene
<i>FNDC3B</i>	1	0.8093	CMC	3' UTR variants	non-EDS gene
<i>LCA5</i>	1	0.8093	CMC	3' UTR variants	non-EDS gene
<i>DOCK9</i>	1	0.8093	CMC	5' UTR variants	non-EDS gene

<i>FASLG</i>	1	0.8093	CMC	5' UTR variants	non-EDS gene
<i>TF</i>	1	0.8093	CMC	5' UTR variants	non-EDS gene
<i>DCAF11</i>	1	0.8093	CMC	Intronic variants	non-EDS gene
<i>LOX</i>	1	0.8093	CMC	Intronic variants	non-EDS gene
<i>LCA5</i>	1	0.8093	CMC	Intronic variants	non-EDS gene
<i>TGFBI</i>	1	0.8093	CMC	Intronic variants	non-EDS gene
<i>SOD1</i>	1	0.8093	CMC	Intronic variants	non-EDS gene
<i>FOXO1</i>	1	0.8093	CMC	Synonymous variants	non-EDS gene
<i>HLA-G</i>	1	0.8093	CMC	Synonymous variants	non-EDS gene
<i>LCA5</i>	1	0.8093	CMC	Synonymous variants	non-EDS gene
<i>RDH13</i>	1	0.8093	CMC	Synonymous variants	non-EDS gene
<i>SOD1</i>	1	0.8093	CMC	Synonymous variants	non-EDS gene
<i>TGFBI</i>	1	0.8093	CMC	Synonymous variants	non-EDS gene

MAF = Minor Allele Frequency; Vt = Variable Thresholds methods; kbac = kernel-based adaptive cluster; CMC = Combined and Multivariate collapsing; skat = SNP-set (Sequence) Kernel Association Method; EDS = Ehlers-Danlos Syndrome. Hg19 was used as reference genome.

**Supplementary table S4 ter:** Results from the gene-based tests, at MAF cutoffs 1% , sorted by significance

<b>Gene</b>	<b>p-value</b>	<b>q-value</b>	<b>Test</b>	<b>Variant category</b>	<b>EDS gene</b>
<i>COL5A1</i>	7.00E-04	0.412	skat	Synonymous variant	EDS gene
<i>MMP9</i>	0.0014	0.4276	kbac	Intronic variants	non-EDS gene
<i>RDH13</i>	0.0038	0.5282	kbac	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.0056	0.5282	vt	Synonymous variant	non-EDS gene
<i>MMP9</i>	0.0067	0.5282	CMC	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.0073	0.5282	skat	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.0078	0.5282	skat	Synonymous variant	non-EDS gene
<i>COL2A1</i>	0.0082	0.5282	vt	Intronic variants	non-EDS gene
<i>MMP9</i>	0.011	0.5282	vt	Intronic variants	non-EDS gene
<i>ZNF469</i>	0.0119	0.5282	skat	Nonsynonymous variants and in-frame indels	EDS gene
<i>COL12A1</i>	0.0123	0.5282	skat	Synonymous variant	EDS gene
<i>DOCK9</i>	0.0126	0.5282	calpha	Synonymous variant	non-EDS gene
<i>RDH13</i>	0.013	0.5282	CMC	Intronic variants	non-EDS gene
<i>FNDC3B</i>	0.0132	0.5282	kbac	Intronic variants	non-EDS gene
<i>ZEB1</i>	0.0144	0.5282	kbac	Synonymous variant	non-EDS gene
<i>DOCK9</i>	0.015	0.5282	kbac	Synonymous variant	non-EDS gene
<i>DOCK9</i>	0.0155	0.5282	CMC	Synonymous variant	non-EDS gene
<i>FNDC3B</i>	0.0189	0.5282	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.0212	0.5282	vt	Intronic variants	non-EDS gene
<i>TF</i>	0.0214	0.5282	calpha	Synonymous variant	non-EDS gene
<i>SOD1</i>	0.0216	0.5282	calpha	5' UTR variants	non-EDS gene
<i>TF</i>	0.0225	0.5282	skat	Synonymous variant	non-EDS gene
<i>LCA5</i>	0.0229	0.5282	skat	Intronic variants	non-EDS gene
<i>COL12A1</i>	0.0238	0.5282	vt	Intronic variants	EDS gene
<i>PARP2</i>	0.0241	0.5282	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.0245	0.5282	skat	Intronic variants	non-EDS gene
<i>P3H4</i>	0.025	0.5282	vt	Synonymous variant	non-EDS gene
<i>FNDC3B</i>	0.0252	0.5282	vt	Intronic variants	non-EDS gene

<i>LCA5</i>	0.0253	0.5282	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FNDC3B</i>	0.028	0.5282	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.0283	0.5282	CMC	Intronic variants	EDS gene
<i>LCA5</i>	0.0286	0.5282	calpha	Intronic variants	non-EDS gene
<i>LCA5</i>	0.0288	0.5282	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.0294	0.5282	calpha	Intronic variants	non-EDS gene
<i>HLA-G</i>	0.031	0.5328	skat	Intronic variants	non-EDS gene
<i>ZEB1</i>	0.0314	0.5328	kbac	Intronic variants	non-EDS gene
<i>FNDC3B</i>	0.0329	0.5439	CMC	Intronic variants	non-EDS gene
<i>PARP2</i>	0.035	0.5627	vt	Synonymous variant	non-EDS gene
<i>ZEB1</i>	0.0364	0.57	CMC	Synonymous variant	non-EDS gene
<i>IL1RN</i>	0.0386	0.5702	kbac	Synonymous variant	non-EDS gene
<i>TF</i>	0.04	0.5702	vt	Synonymous variant	non-EDS gene
<i>IL1RN</i>	0.0404	0.5702	vt	Synonymous variant	non-EDS gene
<i>COL5A1</i>	0.0409	0.5702	skat	Intronic variants	EDS gene
<i>LCA5</i>	0.0422	0.5702	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>ZEB1</i>	0.0428	0.5702	vt	Synonymous variant	non-EDS gene
<i>PARP2</i>	0.043	0.5702	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.0446	0.5702	calpha	Intronic variants	EDS gene
<i>PARP1</i>	0.0448	0.5702	calpha	Synonymous variant	non-EDS gene
<i>LOX</i>	0.05	0.5728	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.0502	0.5728	calpha	Nonsynonymous variants and in-frame indels	EDS gene
<i>TNXB</i>	0.0502	0.5728	calpha	Nonsynonymous variants and in-frame indels	EDS gene
<i>PRPH2</i>	0.0504	0.5728	calpha	Synonymous variant	non-EDS gene
<i>MMP9</i>	0.052	0.5728	skat	Synonymous variant	non-EDS gene
<i>PARP2</i>	0.0538	0.5728	kbac	Synonymous variant	non-EDS gene
<i>ZEB1</i>	0.054	0.5728	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.0545	0.5728	skat	Nonsynonymous variants and in-frame indels	EDS gene
<i>RDH13</i>	0.0558	0.5728	skat	Nonsense and frameshift variants	non-EDS gene
<i>ZEB1</i>	0.0562	0.5728	vt	Intronic variants	non-EDS gene



<i>PARP1</i>	0.0591	0.5728	CMC	Synonymous variant	non-EDS gene
<i>ZEB1</i>	0.0605	0.5728	skat	Synonymous variant	non-EDS gene
<i>COL12A1</i>	0.0606	0.5728	kbac	Synonymous variant	EDS gene
<i>SOD1</i>	0.0628	0.5728	skat	Intronic variants	non-EDS gene
<i>WNT10A</i>	0.0632	0.5728	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>WNT10A</i>	0.0632	0.5728	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>HLA-G</i>	0.0636	0.5728	calpha	Intronic variants	non-EDS gene
<i>DOCK1</i>	0.0648	0.5728	vt	Synonymous variant	non-EDS gene
<i>SOD1</i>	0.0662	0.5728	calpha	Intronic variants	non-EDS gene
<i>LCA5</i>	0.0674	0.5728	CMC	Intronic variants	non-EDS gene
<i>IL1RN</i>	0.068	0.5728	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>P3H4</i>	0.0684	0.5728	kbac	Synonymous variant	non-EDS gene
<i>SLC4A11</i>	0.0688	0.5728	skat	5' UTR variants	non-EDS gene
<i>IL1RN</i>	0.069	0.5728	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL2A1</i>	0.07	0.5728	vt	Synonymous variant	non-EDS gene
<i>PARP1</i>	0.0712	0.5728	skat	Synonymous variant	non-EDS gene
<i>PRPH2</i>	0.0715	0.5728	CMC	Synonymous variant	non-EDS gene
<i>ZEB1</i>	0.0716	0.5728	calpha	Intronic variants	non-EDS gene
<i>RDH13</i>	0.0722	0.5728	calpha	Nonsense and frameshift variants	non-EDS gene
<i>COL12A1</i>	0.0736	0.5764	kbac	Intronic variants	EDS gene
<i>MMP9</i>	0.0762	0.5833	calpha	Intronic variants	non-EDS gene
<i>LOX</i>	0.077	0.5833	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.0793	0.5833	skat	3' UTR variants	non-EDS gene
<i>COL5A1</i>	0.0796	0.5833	calpha	Synonymous variant	EDS gene
<i>VSX1</i>	0.0806	0.5833	vt	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.0846	0.5833	kbac	Intronic variants	non-EDS gene
<i>FNDC3B</i>	0.085	0.5833	vt	Synonymous variant	non-EDS gene
<i>COL2A1</i>	0.086	0.5833	kbac	Intronic variants	non-EDS gene
<i>P3H4</i>	0.087	0.5833	CMC	Synonymous variant	non-EDS gene
<i>IL1RN</i>	0.0888	0.5833	vt	Intronic variants	non-EDS gene

<i>FNDC3B</i>	0.0914	0.5833	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>WNT10A</i>	0.0924	0.5833	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A1</i>	0.0944	0.5833	vt	Intronic variants	EDS gene
<i>IL1RN</i>	0.0962	0.5833	CMC	Synonymous variant	non-EDS gene
<i>PARP2</i>	0.0962	0.5833	CMC	Synonymous variant	non-EDS gene
<i>ZEB1</i>	0.0971	0.5833	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>WNT10A</i>	0.0971	0.5833	skat	5' UTR variants	non-EDS gene
<i>IL1RN</i>	0.0992	0.5833	kbac	Intronic variants	non-EDS gene
<i>TGFBI</i>	0.0992	0.5833	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.0994	0.5833	calpha	5' UTR variants	non-EDS gene
<i>COL12A1</i>	0.1004	0.5833	vt	Synonymous variant	EDS gene
<i>SOD1</i>	0.1012	0.5833	skat	5' UTR variants	non-EDS gene
<i>PARP2</i>	0.1014	0.5833	calpha	Synonymous variant	non-EDS gene
<i>ZEB1</i>	0.1014	0.5833	calpha	Synonymous variant	non-EDS gene
<i>COL12A1</i>	0.1015	0.5833	CMC	Intronic variants	EDS gene
<i>COL2A1</i>	0.1023	0.5833	CMC	Intronic variants	non-EDS gene
<i>SLC4A11</i>	0.1036	0.5833	kbac	5' UTR variants	non-EDS gene
<i>COL12A1</i>	0.1042	0.5833	skat	Nonsynonymous variants and in-frame indels	EDS gene
<i>IL1RN</i>	0.1058	0.5833	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LCA5</i>	0.1058	0.5833	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.1062	0.5833	vt	5' UTR variants	non-EDS gene
<i>FOXO1</i>	0.1076	0.5833	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>MMP9</i>	0.1082	0.5833	skat	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.1088	0.5833	vt	Intronic variants	non-EDS gene
<i>ZEB1</i>	0.1104	0.5833	CMC	Intronic variants	non-EDS gene
<i>TNXB</i>	0.112	0.5833	vt	Nonsynonymous variants and in-frame indels	EDS gene
<i>DOCK1</i>	0.1148	0.5833	skat	5' UTR variants	non-EDS gene
<i>IL1RN</i>	0.115	0.5833	skat	Intronic variants	non-EDS gene
<i>VSX1</i>	0.116	0.5833	kbac	Intronic variants	non-EDS gene
<i>LOX</i>	0.1174	0.5833	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene

<i>LOX</i>	0.1174	0.5833	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.1184	0.5833	CMC	Synonymous variant	non-EDS gene
<i>TF</i>	0.1191	0.5833	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FNDC3B</i>	0.1209	0.5833	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.1214	0.5833	skat	3' UTR variants	non-EDS gene
<i>HLA-G</i>	0.1219	0.5833	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>HLA-G</i>	0.1219	0.5833	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.1219	0.5833	kbac	Synonymous variant	non-EDS gene
<i>ZEB1</i>	0.1239	0.5833	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.1241	0.5833	skat	3' UTR variants	non-EDS gene
<i>DCAF11</i>	0.1249	0.5833	kbac	3' UTR variants	non-EDS gene
<i>COL12A1</i>	0.1249	0.5833	skat	Intronic variants	EDS gene
<i>MIR184</i>	0.1268	0.5833	skat	noncoding RNA 5'	non-EDS gene
<i>DOCK9</i>	0.1269	0.5833	calpha	Intronic variants	non-EDS gene
<i>HLA-G</i>	0.127	0.5833	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.1289	0.5876	kbac	3' UTR variants	non-EDS gene
<i>TNXB</i>	0.1329	0.6013	calpha	Synonymous variant	EDS gene
<i>MMP9</i>	0.1359	0.6104	kbac	Synonymous variant	non-EDS gene
<i>VSX1</i>	0.1369	0.6104	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.1383	0.6123	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A1</i>	0.1419	0.6227	calpha	Intronic variants	EDS gene
<i>ZEB1</i>	0.1427	0.6227	skat	Intronic variants	non-EDS gene
<i>TF</i>	0.1439	0.6234	kbac	Synonymous variant	non-EDS gene
<i>FNDC3B</i>	0.1456	0.6265	skat	Intronic variants	non-EDS gene
<i>RDH13</i>	0.1498	0.6383	skat	5' UTR variants	non-EDS gene
<i>TNXB</i>	0.1504	0.6383	skat	Nonsense and frameshift variants	EDS gene
<i>RDH13</i>	0.1518	0.6399	vt	Nonsense and frameshift variants	non-EDS gene
<i>NLRP1</i>	0.1548	0.6467	calpha	3' UTR variants	non-EDS gene
<i>WNT10A</i>	0.1556	0.6467	skat	Synonymous variant	non-EDS gene
<i>PARP1</i>	0.1568	0.6475	calpha	Nonsense and frameshift variants	non-EDS gene

<i>COL5A1</i>	0.1588	0.6514	kbac	Intronic variants	EDS gene
<i>IL1RN</i>	0.1628	0.654	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>IL1RN</i>	0.1628	0.654	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.1636	0.654	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.1638	0.654	skat	Synonymous variant	EDS gene
<i>HLA-G</i>	0.1668	0.6573	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP2</i>	0.1678	0.6573	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP2</i>	0.1678	0.6573	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.1708	0.661	kbac	Nonsynonymous variants and in-frame indels	EDS gene
<i>FOXO1</i>	0.1718	0.661	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DCAF11</i>	0.172	0.661	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FOXO1</i>	0.1788	0.6649	calpha	Synonymous variant	non-EDS gene
<i>MMP9</i>	0.1798	0.6649	vt	Synonymous variant	non-EDS gene
<i>PARP1</i>	0.1804	0.6649	skat	Intronic variants	non-EDS gene
<i>COL2A1</i>	0.1818	0.6649	kbac	Synonymous variant	non-EDS gene
<i>HLA-G</i>	0.1838	0.6649	vt	Intronic variants	non-EDS gene
<i>ZEB1</i>	0.1842	0.6649	skat	Nonsense and frameshift variants	non-EDS gene
<i>IL1RN</i>	0.1843	0.6649	skat	5' UTR variants	non-EDS gene
<i>VSX1</i>	0.1853	0.6649	skat	Nonsense and frameshift variants	non-EDS gene
<i>IL1RN</i>	0.1858	0.6649	calpha	Intronic variants	non-EDS gene
<i>FASLG</i>	0.1863	0.6649	skat	Intronic variants	non-EDS gene
<i>COL12A1</i>	0.1864	0.6649	skat	Nonsense and frameshift variants	EDS gene
<i>LAMC3</i>	0.1868	0.6649	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FASLG</i>	0.1876	0.6649	skat	Synonymous variant	non-EDS gene
<i>COL5A3</i>	0.1928	0.6649	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.1928	0.6649	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LCA5</i>	0.1938	0.6649	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LCA5</i>	0.1938	0.6649	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK9</i>	0.1941	0.6649	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DCAF11</i>	0.1948	0.6649	kbac	Synonymous variant	non-EDS gene

<i>P3H4</i>	0.1948	0.6649	vt	Intronic variants	non-EDS gene
<i>PARP2</i>	0.1997	0.6741	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>WNT10A</i>	0.1998	0.6741	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.2008	0.6741	kbac	Synonymous variant	non-EDS gene
<i>SLC4A11</i>	0.2029	0.6776	CMC	5' UTR variants	non-EDS gene
<i>P3H4</i>	0.2048	0.6801	calpha	Synonymous variant	non-EDS gene
<i>FOXO1</i>	0.2089	0.6855	skat	Synonymous variant	non-EDS gene
<i>NLRP1</i>	0.2094	0.6855	skat	Nonsense and frameshift variants	non-EDS gene
<i>FOXO1</i>	0.2098	0.6855	kbac	Synonymous variant	non-EDS gene
<i>COL12A1</i>	0.2228	0.7196	calpha	Intronic variants	EDS gene
<i>IL1RN</i>	0.2228	0.7196	calpha	Synonymous variant	non-EDS gene
<i>DOCK1</i>	0.2238	0.7196	vt	Intronic variants	non-EDS gene
<i>COL5A3</i>	0.23	0.7357	CMC	3' UTR variants	non-EDS gene
<i>MIR184</i>	0.233	0.7413	CMC	noncoding RNA 5'	non-EDS gene
<i>FNDC3B</i>	0.2358	0.7425	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FNDC3B</i>	0.2358	0.7425	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FOXO1</i>	0.2378	0.745	vt	Synonymous variant	non-EDS gene
<i>RDH13</i>	0.2398	0.746	kbac	Nonsense and frameshift variants	non-EDS gene
<i>TNXB</i>	0.2414	0.746	CMC	Synonymous variant	EDS gene
<i>VSX1</i>	0.2418	0.746	calpha	Intronic variants	non-EDS gene
<i>VSX1</i>	0.2465	0.7476	skat	3' UTR variants	non-EDS gene
<i>TF</i>	0.2468	0.7476	vt	Intronic variants	non-EDS gene
<i>P3H4</i>	0.2488	0.7476	kbac	Intronic variants	non-EDS gene
<i>DOCK1</i>	0.2498	0.7476	kbac	Intronic variants	non-EDS gene
<i>FOXO1</i>	0.2507	0.7476	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FOXO1</i>	0.2507	0.7476	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.2508	0.7476	CMC	Nonsense and frameshift variants	non-EDS gene
<i>MMP9</i>	0.2557	0.7585	calpha	Synonymous variant	non-EDS gene
<i>DCAF11</i>	0.2577	0.7594	calpha	3' UTR variants	non-EDS gene
<i>IL1RN</i>	0.2586	0.7594	skat	Synonymous variant	non-EDS gene

<i>WNT10A</i>	0.2598	0.7594	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A1</i>	0.2627	0.7641	kbac	Synonymous variant	EDS gene
<i>COL2A1</i>	0.2646	0.7641	CMC	Synonymous variant	non-EDS gene
<i>DCAF11</i>	0.2667	0.7641	calpha	Intronic variants	non-EDS gene
<i>VSX1</i>	0.267	0.7641	skat	Intronic variants	non-EDS gene
<i>DCAF11</i>	0.2687	0.7641	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FOXO1</i>	0.2701	0.7641	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>IL1RN</i>	0.2701	0.7641	CMC	Intronic variants	non-EDS gene
<i>PARP2</i>	0.2737	0.7699	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP2</i>	0.2767	0.7699	kbac	Intronic variants	non-EDS gene
<i>DCAF11</i>	0.2786	0.7699	skat	3' UTR variants	non-EDS gene
<i>MMP9</i>	0.2797	0.7699	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>MMP9</i>	0.2797	0.7699	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DCAF11</i>	0.2797	0.7699	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL12A1</i>	0.2887	0.77	calpha	Synonymous variant	EDS gene
<i>DCAF11</i>	0.2887	0.77	kbac	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.2897	0.77	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK9</i>	0.2897	0.77	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>ZNF469</i>	0.2897	0.77	calpha	Synonymous variant	EDS gene
<i>PARP1</i>	0.2897	0.77	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FNDC3B</i>	0.2917	0.77	kbac	Synonymous variant	non-EDS gene
<i>COL12A1</i>	0.2926	0.77	CMC	Synonymous variant	EDS gene
<i>LOXHD1</i>	0.2927	0.77	vt	Synonymous variant	non-EDS gene
<i>DCAF11</i>	0.293	0.77	CMC	3' UTR variants	non-EDS gene
<i>SOD1</i>	0.2936	0.77	CMC	5' UTR variants	non-EDS gene
<i>ZEB1</i>	0.3007	0.7818	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>ZEB1</i>	0.3007	0.7818	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.3078	0.797	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DCAF11</i>	0.3107	0.801	vt	3' UTR variants	non-EDS gene
<i>MMP9</i>	0.3127	0.8027	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene

<i>COL5A3</i>	0.3217	0.8224	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.3257	0.8291	calpha	Nonsense and frameshift variants	non-EDS gene
<i>HLA-G</i>	0.3357	0.851	kbac	Synonymous variant	non-EDS gene
<i>P3H4</i>	0.3392	0.854	skat	Synonymous variant	non-EDS gene
<i>TGFBI</i>	0.3397	0.854	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.3476	0.8703	skat	Synonymous variant	non-EDS gene
<i>COL2A1</i>	0.3499	0.8726	skat	Synonymous variant	non-EDS gene
<i>LOXHD1</i>	0.3526	0.8744	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.3536	0.8744	calpha	Synonymous variant	non-EDS gene
<i>MMP9</i>	0.3549	0.8744	CMC	Synonymous variant	non-EDS gene
<i>DCAF11</i>	0.3579	0.8768	skat	Intronic variants	non-EDS gene
<i>PARP1</i>	0.3596	0.8768	calpha	Intronic variants	non-EDS gene
<i>COL2A1</i>	0.3606	0.8768	calpha	Intronic variants	non-EDS gene
<i>HLA-G</i>	0.3616	0.8768	kbac	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.3646	0.8795	calpha	3' UTR variants	non-EDS gene
<i>TF</i>	0.3656	0.8795	kbac	Intronic variants	non-EDS gene
<i>WNT10A</i>	0.3685	0.8798	CMC	5' UTR variants	non-EDS gene
<i>DCAF11</i>	0.3686	0.8798	vt	Synonymous variant	non-EDS gene
<i>TF</i>	0.374	0.8892	skat	Intronic variants	non-EDS gene
<i>LAMC3</i>	0.3766	0.8919	kbac	Intronic variants	non-EDS gene
<i>LOX</i>	0.3836	0.9	calpha	3' UTR variants	non-EDS gene
<i>NLRP1</i>	0.3836	0.9	calpha	Synonymous variant	non-EDS gene
<i>DCAF11</i>	0.3856	0.9	vt	Intronic variants	non-EDS gene
<i>SLC4A11</i>	0.3893	0.9	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>WNT10A</i>	0.3896	0.9	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FNDC3B</i>	0.3906	0.9	calpha	Intronic variants	non-EDS gene
<i>LOXHD1</i>	0.394	0.9	skat	5' UTR variants	non-EDS gene
<i>DOCK9</i>	0.3976	0.9	calpha	3' UTR variants	non-EDS gene
<i>LAMC3</i>	0.3978	0.9	skat	Synonymous variant	non-EDS gene
<i>PRPH2</i>	0.3986	0.9	calpha	Intronic variants	non-EDS gene

<i>COL5A3</i>	0.3986	0.9	vt	Synonymous variant	non-EDS gene
<i>PARP2</i>	0.4001	0.9	CMC	Intronic variants	non-EDS gene
<i>LOXHD1</i>	0.4006	0.9	vt	Nonsense and frameshift variants	non-EDS gene
<i>DOCK1</i>	0.4017	0.9	CMC	Intronic variants	non-EDS gene
<i>IL1RN</i>	0.4036	0.9	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.4036	0.9	CMC	5' UTR variants	non-EDS gene
<i>RDH13</i>	0.4108	0.9075	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>EPPK1</i>	0.4194	0.9075	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.4206	0.9075	kbac	Intronic variants	non-EDS gene
<i>COL5A1</i>	0.4212	0.9075	CMC	Intronic variants	EDS gene
<i>SLC4A11</i>	0.4256	0.9075	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.4256	0.9075	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.4266	0.9075	kbac	Nonsense and frameshift variants	non-EDS gene
<i>TF</i>	0.4276	0.9075	calpha	Intronic variants	non-EDS gene
<i>HLA-G</i>	0.4276	0.9075	CMC	Intronic variants	non-EDS gene
<i>EPPK1</i>	0.4307	0.9075	CMC	Synonymous variant	non-EDS gene
<i>P3H4</i>	0.4326	0.9075	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>P3H4</i>	0.4326	0.9075	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.4348	0.9075	CMC	3' UTR variants	non-EDS gene
<i>DCAF11</i>	0.4352	0.9075	skat	Synonymous variant	non-EDS gene
<i>SLC4A11</i>	0.4366	0.9075	vt	Intronic variants	non-EDS gene
<i>VSX1</i>	0.4376	0.9075	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LAMC3</i>	0.4406	0.9075	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.4436	0.9075	kbac	Intronic variants	non-EDS gene
<i>ZNF469</i>	0.4436	0.9075	kbac	Synonymous variant	EDS gene
<i>ZNF469</i>	0.4505	0.9075	calpha	Nonsynonymous variants and in-frame indels	EDS gene
<i>ZNF469</i>	0.4505	0.9075	calpha	Nonsynonymous variants and in-frame indels	EDS gene
<i>COL2A1</i>	0.4525	0.9075	calpha	Synonymous variant	non-EDS gene
<i>DCAF11</i>	0.4565	0.9075	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL12A1</i>	0.4575	0.9075	calpha	Nonsynonymous variants and in-frame indels	EDS gene



<i>COL12A1</i>	0.4575	0.9075	calpha	Nonsynonymous variants and in-frame indels	EDS gene
<i>RDH13</i>	0.4583	0.9075	CMC	5' UTR variants	non-EDS gene
<i>WNT10A</i>	0.4608	0.9075	CMC	Synonymous variant	non-EDS gene
<i>FASLG</i>	0.4615	0.9075	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.4615	0.9075	kbac	Nonsense and frameshift variants	EDS gene
<i>NLRP1</i>	0.4648	0.9075	skat	Synonymous variant	non-EDS gene
<i>SOD1</i>	0.4685	0.9075	vt	Intronic variants	non-EDS gene
<i>FASLG</i>	0.4695	0.9075	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.4705	0.9075	vt	Nonsense and frameshift variants	EDS gene
<i>SLC4A11</i>	0.4705	0.9075	vt	Synonymous variant	non-EDS gene
<i>DCAF11</i>	0.4715	0.9075	CMC	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.4723	0.9075	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LCA5</i>	0.4735	0.9075	vt	Synonymous variant	non-EDS gene
<i>IL1RN</i>	0.475	0.9075	CMC	5' UTR variants	non-EDS gene
<i>DOCK1</i>	0.4752	0.9075	skat	Intronic variants	non-EDS gene
<i>TGFBI</i>	0.4765	0.9075	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TGFBI</i>	0.4765	0.9075	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LAMC3</i>	0.4775	0.9075	kbac	Synonymous variant	non-EDS gene
<i>COL5A1</i>	0.4785	0.9075	vt	Synonymous variant	EDS gene
<i>ZEB1</i>	0.4791	0.9075	CMC	Nonsense and frameshift variants	non-EDS gene
<i>VSX1</i>	0.4794	0.9075	CMC	Nonsense and frameshift variants	non-EDS gene
<i>COL12A1</i>	0.4797	0.9075	CMC	Nonsense and frameshift variants	EDS gene
<i>FASLG</i>	0.48	0.9075	CMC	Synonymous variant	non-EDS gene
<i>COL5A3</i>	0.4815	0.9075	calpha	Synonymous variant	non-EDS gene
<i>SOD1</i>	0.4835	0.9075	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.4844	0.9075	CMC	Nonsense and frameshift variants	non-EDS gene
<i>VSX1</i>	0.4887	0.9075	CMC	3' UTR variants	non-EDS gene
<i>VSX1</i>	0.4896	0.9075	CMC	Intronic variants	non-EDS gene
<i>P3H4</i>	0.491	0.9075	CMC	Intronic variants	non-EDS gene
<i>DCAF11</i>	0.4912	0.9075	CMC	Synonymous variant	non-EDS gene

<i>LAMC3</i>	0.4942	0.9075	skat	Intronic variants	non-EDS gene
<i>SOD1</i>	0.4965	0.9075	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.4995	0.9075	calpha	5' UTR variants	non-EDS gene
<i>PRPH2</i>	0.4997	0.9075	CMC	Intronic variants	non-EDS gene
<i>FASLG</i>	0.5001	0.9075	CMC	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.5003	0.9075	CMC	3' UTR variants	non-EDS gene
<i>COL5A3</i>	0.5005	0.9075	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.5006	0.9075	CMC	3' UTR variants	non-EDS gene
<i>SLC4A11</i>	0.5014	0.9075	skat	Synonymous variant	non-EDS gene
<i>LOX</i>	0.5025	0.9075	calpha	Synonymous variant	non-EDS gene
<i>FNDC3B</i>	0.5035	0.9075	calpha	Synonymous variant	non-EDS gene
<i>DOCK1</i>	0.5095	0.9109	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.5095	0.9109	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>P3H4</i>	0.51	0.9109	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.5115	0.9109	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.5135	0.9109	kbac	Synonymous variant	non-EDS gene
<i>RDH13</i>	0.5144	0.9109	skat	Synonymous variant	non-EDS gene
<i>VSX1</i>	0.5225	0.9194	kbac	Synonymous variant	non-EDS gene
<i>COL12A1</i>	0.5235	0.9194	kbac	Nonsynonymous variants and in-frame indels	EDS gene
<i>TF</i>	0.5236	0.9194	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP2</i>	0.5315	0.9282	vt	Intronic variants	non-EDS gene
<i>COL12A1</i>	0.5335	0.9282	vt	Nonsynonymous variants and in-frame indels	EDS gene
<i>COL2A1</i>	0.5345	0.9282	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.5355	0.9282	vt	3' UTR variants	non-EDS gene
<i>VSX1</i>	0.5365	0.9282	vt	Synonymous variant	non-EDS gene
<i>SLC4A11</i>	0.5378	0.9282	CMC	Nonsense and frameshift variants	non-EDS gene
<i>COL12A1</i>	0.55	0.9392	CMC	Nonsynonymous variants and in-frame indels	EDS gene
<i>LAMC3</i>	0.5514	0.9392	vt	Intronic variants	non-EDS gene
<i>FASLG</i>	0.5536	0.9392	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.5544	0.9392	kbac	Intronic variants	non-EDS gene

<i>SLC4A11</i>	0.5554	0.9392	kbac	Synonymous variant	non-EDS gene
<i>VSX1</i>	0.5556	0.9392	skat	Synonymous variant	non-EDS gene
<i>TF</i>	0.5564	0.9392	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TF</i>	0.5564	0.9392	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FNDC3B</i>	0.5582	0.9396	skat	Synonymous variant	non-EDS gene
<i>DOCK1</i>	0.5654	0.9473	calpha	Intronic variants	non-EDS gene
<i>PARP1</i>	0.5674	0.9473	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.5674	0.9473	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>HLA-G</i>	0.5724	0.953	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.5755	0.9555	skat	Nonsense and frameshift variants	non-EDS gene
<i>DOCK1</i>	0.5784	0.9578	vt	3' UTR variants	non-EDS gene
<i>ZNF469</i>	0.5815	0.9586	CMC	Nonsynonymous variants and in-frame indels	EDS gene
<i>DOCK1</i>	0.5821	0.9586	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SOD1</i>	0.5873	0.9596	CMC	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.5891	0.9596	skat	3' UTR variants	non-EDS gene
<i>LCA5</i>	0.5914	0.9596	kbac	Synonymous variant	non-EDS gene
<i>LOXHD1</i>	0.5914	0.9596	skat	Synonymous variant	non-EDS gene
<i>COL2A1</i>	0.5944	0.9596	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.5947	0.9596	CMC	Synonymous variant	non-EDS gene
<i>LOXHD1</i>	0.5959	0.9596	CMC	Intronic variants	non-EDS gene
<i>COL5A1</i>	0.5991	0.9596	CMC	Synonymous variant	EDS gene
<i>DOCK1</i>	0.6014	0.9596	kbac	3' UTR variants	non-EDS gene
<i>NLRP1</i>	0.6054	0.9596	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.6054	0.9596	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.6114	0.9596	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>EPPK1</i>	0.6124	0.9596	vt	Nonsense and frameshift variants	non-EDS gene
<i>HLA-G</i>	0.6127	0.9596	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.6134	0.9596	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.6134	0.9596	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK9</i>	0.6134	0.9596	kbac	3' UTR variants	non-EDS gene

<i>LAMC3</i>	0.6144	0.9596	calpha	Intronic variants	non-EDS gene
<i>RDH13</i>	0.6154	0.9596	kbac	Synonymous variant	non-EDS gene
<i>LAMC3</i>	0.6167	0.9596	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.6183	0.9596	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>NLRP1</i>	0.6184	0.9596	vt	Intronic variants	non-EDS gene
<i>FASLG</i>	0.6229	0.9596	skat	5' UTR variants	non-EDS gene
<i>LOXHD1</i>	0.6239	0.9596	CMC	5' UTR variants	non-EDS gene
<i>LOX</i>	0.6257	0.9596	CMC	5' UTR variants	non-EDS gene
<i>LOX</i>	0.6258	0.9596	CMC	Synonymous variant	non-EDS gene
<i>PARP2</i>	0.6263	0.9596	skat	Intronic variants	non-EDS gene
<i>COL5A3</i>	0.6294	0.9596	vt	Intronic variants	non-EDS gene
<i>ZNF469</i>	0.6304	0.9596	kbac	Nonsynonymous variants and in-frame indels	EDS gene
<i>DOCK1</i>	0.6314	0.9596	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK9</i>	0.6314	0.9596	vt	3' UTR variants	non-EDS gene
<i>TF</i>	0.6361	0.9644	skat	5' UTR variants	non-EDS gene
<i>MMP9</i>	0.6394	0.967	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.6427	0.9676	CMC	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.643	0.9676	skat	Intronic variants	non-EDS gene
<i>COL5A1</i>	0.6504	0.9757	kbac	Nonsynonymous variants and in-frame indels	EDS gene
<i>PARP2</i>	0.6529	0.9757	skat	Synonymous variant	non-EDS gene
<i>SLC4A11</i>	0.6533	0.9757	calpha	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.6547	0.9757	skat	5' UTR variants	non-EDS gene
<i>LAMC3</i>	0.6573	0.9772	vt	Synonymous variant	non-EDS gene
<i>ZEB1</i>	0.6599	0.9786	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.6628	0.9806	skat	Intronic variants	non-EDS gene
<i>FNDC3B</i>	0.6661	0.9828	CMC	Synonymous variant	non-EDS gene
<i>TGFBI</i>	0.6683	0.9828	skat	Synonymous variant	non-EDS gene
<i>LAMC3</i>	0.6693	0.9828	calpha	Synonymous variant	non-EDS gene
<i>TNXB</i>	0.6722	0.9828	CMC	Nonsynonymous variants and in-frame indels	EDS gene
<i>PARP1</i>	0.6723	0.9828	vt	Intronic variants	non-EDS gene

<i>PRPH2</i>	0.6753	0.9839	CMC	5' UTR variants	non-EDS gene
<i>P3H4</i>	0.6763	0.9839	calpha	Intronic variants	non-EDS gene
<i>TGFBI</i>	0.6814	0.9863	CMC	Synonymous variant	non-EDS gene
<i>LOX</i>	0.6833	0.9863	vt	Intronic variants	non-EDS gene
<i>LOXHD1</i>	0.6853	0.9863	calpha	Synonymous variant	non-EDS gene
<i>RDH13</i>	0.6873	0.9863	vt	Synonymous variant	non-EDS gene
<i>ZNF469</i>	0.6883	0.9863	vt	Synonymous variant	EDS gene
<i>LOX</i>	0.6893	0.9863	kbac	Intronic variants	non-EDS gene
<i>COL5A1</i>	0.6933	0.9863	skat	Nonsynonymous variants and in-frame indels	EDS gene
<i>LOXHD1</i>	0.6953	0.9863	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.6953	0.9863	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>VSX1</i>	0.6956	0.9863	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>ZNF469</i>	0.6963	0.9863	vt	Nonsynonymous variants and in-frame indels	EDS gene
<i>P3H4</i>	0.7006	0.9863	skat	Intronic variants	non-EDS gene
<i>COL5A3</i>	0.7011	0.9863	skat	Intronic variants	non-EDS gene
<i>ZNF469</i>	0.7043	0.9863	CMC	Synonymous variant	EDS gene
<i>EPPK1</i>	0.7083	0.9863	kbac	Nonsense and frameshift variants	non-EDS gene
<i>HLA-G</i>	0.7083	0.9863	vt	Synonymous variant	non-EDS gene
<i>SOD1</i>	0.7092	0.9863	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.7096	0.9863	skat	Intronic variants	non-EDS gene
<i>NLRP1</i>	0.7103	0.9863	kbac	Synonymous variant	non-EDS gene
<i>TGFBI</i>	0.7123	0.9863	kbac	Intronic variants	non-EDS gene
<i>PARP1</i>	0.7133	0.9863	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TGFBI</i>	0.7143	0.9863	vt	Intronic variants	non-EDS gene
<i>VSX1</i>	0.7183	0.9863	calpha	Synonymous variant	non-EDS gene
<i>DOCK1</i>	0.7187	0.9863	skat	Synonymous variant	non-EDS gene
<i>NLRP1</i>	0.7219	0.9863	CMC	Synonymous variant	non-EDS gene
<i>P3H4</i>	0.7223	0.9863	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SOD1</i>	0.7293	0.9863	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SOD1</i>	0.7293	0.9863	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene

<i>TNXB</i>	0.7323	0.9863	calpha	Nonsense and frameshift variants	EDS gene
<i>DOCK1</i>	0.7323	0.9863	calpha	Synonymous variant	non-EDS gene
<i>LOXHD1</i>	0.7343	0.9863	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK1</i>	0.7348	0.9863	skat	3' UTR variants	non-EDS gene
<i>DCAF11</i>	0.7363	0.9863	calpha	Synonymous variant	non-EDS gene
<i>LCA5</i>	0.7393	0.9863	calpha	Synonymous variant	non-EDS gene
<i>NLRP1</i>	0.7393	0.9863	kbac	Intronic variants	non-EDS gene
<i>TGFBI</i>	0.7393	0.9863	vt	Synonymous variant	non-EDS gene
<i>LOX</i>	0.7398	0.9863	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FASLG</i>	0.7413	0.9863	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.7447	0.9863	CMC	Nonsense and frameshift variants	non-EDS gene
<i>TGFBI</i>	0.7463	0.9863	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>DOCK9</i>	0.7473	0.9863	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.7473	0.9863	kbac	5' UTR variants	non-EDS gene
<i>PRPH2</i>	0.7553	0.9863	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL2A1</i>	0.7557	0.9863	skat	Intronic variants	non-EDS gene
<i>DCAF11</i>	0.7564	0.9863	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LAMC3</i>	0.7601	0.9863	CMC	Intronic variants	non-EDS gene
<i>SOD1</i>	0.7632	0.9863	kbac	Intronic variants	non-EDS gene
<i>COL5A1</i>	0.7642	0.9863	vt	Nonsynonymous variants and in-frame indels	EDS gene
<i>P3H4</i>	0.7686	0.9863	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LCA5</i>	0.7707	0.9863	skat	Synonymous variant	non-EDS gene
<i>LCA5</i>	0.7722	0.9863	kbac	Intronic variants	non-EDS gene
<i>LOX</i>	0.7732	0.9863	vt	Synonymous variant	non-EDS gene
<i>TNXB</i>	0.7732	0.9863	vt	Synonymous variant	EDS gene
<i>LOXHD1</i>	0.7738	0.9863	skat	Nonsense and frameshift variants	non-EDS gene
<i>LOX</i>	0.7749	0.9863	skat	5' UTR variants	non-EDS gene
<i>LOXHD1</i>	0.7762	0.9863	kbac	Intronic variants	non-EDS gene
<i>PARP2</i>	0.7782	0.9863	calpha	Intronic variants	non-EDS gene
<i>DOCK9</i>	0.7791	0.9863	skat	5' UTR variants	non-EDS gene

<i>EPPK1</i>	0.7842	0.9863	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>EPPK1</i>	0.7842	0.9863	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	0.7842	0.9863	calpha	Intronic variants	non-EDS gene
<i>LCA5</i>	0.7862	0.9863	vt	Intronic variants	non-EDS gene
<i>TGFBI</i>	0.7872	0.9863	calpha	Synonymous variant	non-EDS gene
<i>NLRP1</i>	0.7872	0.9863	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.7912	0.9863	kbac	Synonymous variant	non-EDS gene
<i>FNDC3B</i>	0.7922	0.9863	skat	3' UTR variants	non-EDS gene
<i>SOD1</i>	0.7922	0.9863	skat	Synonymous variant	non-EDS gene
<i>COL2A1</i>	0.7935	0.9863	skat	Nonsense and frameshift variants	non-EDS gene
<i>HLA-G</i>	0.7939	0.9863	skat	Synonymous variant	non-EDS gene
<i>LOX</i>	0.7942	0.9863	vt	5' UTR variants	non-EDS gene
<i>P3H4</i>	0.7972	0.9863	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.7972	0.9863	kbac	Nonsense and frameshift variants	non-EDS gene
<i>EPPK1</i>	0.7972	0.9863	vt	Synonymous variant	non-EDS gene
<i>LCA5</i>	0.7984	0.9863	skat	3' UTR variants	non-EDS gene
<i>LOXHD1</i>	0.8002	0.9863	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.8007	0.9863	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.8062	0.9904	vt	Nonsense and frameshift variants	non-EDS gene
<i>MMP9</i>	0.8086	0.9904	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>EPPK1</i>	0.8142	0.9904	skat	Nonsense and frameshift variants	non-EDS gene
<i>COL2A1</i>	0.815	0.9904	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.8158	0.9904	skat	Nonsense and frameshift variants	non-EDS gene
<i>LOXHD1</i>	0.8162	0.9904	calpha	Nonsense and frameshift variants	non-EDS gene
<i>VSX1</i>	0.8182	0.9904	CMC	Synonymous variant	non-EDS gene
<i>LOXHD1</i>	0.8182	0.9904	vt	Intronic variants	non-EDS gene
<i>FOXO1</i>	0.8185	0.9904	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PRPH2</i>	0.8252	0.9944	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PRPH2</i>	0.8252	0.9944	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.8336	1	skat	Nonsynonymous variants and in-frame indels	non-EDS gene

<i>NLRP1</i>	0.8444	1	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TGFBI</i>	0.8462	1	kbac	Synonymous variant	non-EDS gene
<i>TNXB</i>	0.8472	1	kbac	Synonymous variant	EDS gene
<i>EPPK1</i>	0.8499	1	skat	Synonymous variant	non-EDS gene
<i>NLRP1</i>	0.8551	1	vt	Synonymous variant	non-EDS gene
<i>PRPH2</i>	0.8581	1	kbac	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.8619	1	skat	3' UTR variants	non-EDS gene
<i>FOXO1</i>	0.8631	1	CMC	Synonymous variant	non-EDS gene
<i>TNXB</i>	0.8647	1	skat	Intronic variants	EDS gene
<i>PRPH2</i>	0.8681	1	kbac	3' UTR variants	non-EDS gene
<i>TGFBI</i>	0.8683	1	skat	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.8711	1	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A1</i>	0.8751	1	calpha	Nonsynonymous variants and in-frame indels	EDS gene
<i>ZNF469</i>	0.8752	1	skat	Synonymous variant	EDS gene
<i>NLRP1</i>	0.8752	1	CMC	Intronic variants	non-EDS gene
<i>LOX</i>	0.8754	1	skat	3' UTR variants	non-EDS gene
<i>SOD1</i>	0.8761	1	kbac	5' UTR variants	non-EDS gene
<i>PRPH2</i>	0.8761	1	vt	3' UTR variants	non-EDS gene
<i>NLRP1</i>	0.8771	1	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOX</i>	0.8801	1	kbac	3' UTR variants	non-EDS gene
<i>LOX</i>	0.8801	1	vt	3' UTR variants	non-EDS gene
<i>SLC4A11</i>	0.881	1	CMC	Synonymous variant	non-EDS gene
<i>PRPH2</i>	0.8821	1	vt	Intronic variants	non-EDS gene
<i>EPPK1</i>	0.8841	1	calpha	Synonymous variant	non-EDS gene
<i>PARP1</i>	0.8881	1	kbac	Synonymous variant	non-EDS gene
<i>DOCK9</i>	0.8881	1	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SLC4A11</i>	0.889	1	CMC	Intronic variants	non-EDS gene
<i>LOX</i>	0.8931	1	skat	Synonymous variant	non-EDS gene
<i>PARP1</i>	0.8941	1	vt	Synonymous variant	non-EDS gene
<i>PARP1</i>	0.9	1	skat	Nonsense and frameshift variants	non-EDS gene



<i>SLC4A11</i>	0.9001	1	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PRPH2</i>	0.901	1	skat	Synonymous variant	non-EDS gene
<i>LOX</i>	0.9031	1	calpha	Intronic variants	non-EDS gene
<i>EPPK1</i>	0.9031	1	kbac	Synonymous variant	non-EDS gene
<i>PRPH2</i>	0.9051	1	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.9051	1	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>VSX1</i>	0.9061	1	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>VSX1</i>	0.9061	1	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TGFBI</i>	0.9064	1	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>HLA-G</i>	0.9101	1	calpha	Synonymous variant	non-EDS gene
<i>DOCK1</i>	0.9141	1	calpha	3' UTR variants	non-EDS gene
<i>COL5A1</i>	0.9143	1	CMC	Nonsynonymous variants and in-frame indels	EDS gene
<i>EPPK1</i>	0.9161	1	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LOXHD1</i>	0.9196	1	CMC	Synonymous variant	non-EDS gene
<i>VSX1</i>	0.9199	1	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PRPH2</i>	0.9223	1	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.9226	1	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.9311	1	vt	Nonsense and frameshift variants	non-EDS gene
<i>EPPK1</i>	0.9361	1	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	0.9361	1	kbac	Nonsense and frameshift variants	non-EDS gene
<i>SLC4A11</i>	0.9361	1	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>RDH13</i>	0.9391	1	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TF</i>	0.9401	1	vt	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>EPPK1</i>	0.9421	1	calpha	Nonsense and frameshift variants	non-EDS gene
<i>COL2A1</i>	0.954	1	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SOD1</i>	0.956	1	vt	5' UTR variants	non-EDS gene
<i>SLC4A11</i>	0.957	1	calpha	Synonymous variant	non-EDS gene
<i>LOXHD1</i>	0.9728	1	skat	Intronic variants	non-EDS gene
<i>TGFBI</i>	0.974	1	calpha	Intronic variants	non-EDS gene
<i>PRPH2</i>	0.978	1	kbac	Synonymous variant	non-EDS gene

<i>TF</i>	0.988	1	kbac	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PRPH2</i>	0.99	1	vt	Synonymous variant	non-EDS gene
<i>NLRP1</i>	0.991	1	calpha	Intronic variants	non-EDS gene
<i>LOXHD1</i>	0.995	1	calpha	Intronic variants	non-EDS gene
<i>TNXB</i>	0.995	1	vt	Intronic variants	EDS gene
<i>LAMC3</i>	0.996	1	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LAMC3</i>	0.996	1	calpha	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>TNXB</i>	0.997	1	kbac	Intronic variants	EDS gene
<i>COL2A1</i>	1	1	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>FASLG</i>	1	1	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL5A3</i>	1	1	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>MMP9</i>	1	1	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>PARP1</i>	1	1	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>SOD1</i>	1	1	CMC	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL2A1</i>	1	1	CMC	Nonsense and frameshift variants	non-EDS gene
<i>EPPK1</i>	1	1	CMC	Nonsense and frameshift variants	non-EDS gene
<i>RDH13</i>	1	1	CMC	Nonsense and frameshift variants	non-EDS gene
<i>LOX</i>	1	1	CMC	Nonsense and frameshift variants	non-EDS gene
<i>TNXB</i>	1	1	CMC	Nonsense and frameshift variants	EDS gene
<i>COL2A1</i>	1	1	CMC	3' UTR variants	non-EDS gene
<i>DOCK1</i>	1	1	CMC	3' UTR variants	non-EDS gene
<i>FNDC3B</i>	1	1	CMC	3' UTR variants	non-EDS gene
<i>DOCK9</i>	1	1	CMC	3' UTR variants	non-EDS gene
<i>LCA5</i>	1	1	CMC	3' UTR variants	non-EDS gene
<i>SLC4A11</i>	1	1	CMC	3' UTR variants	non-EDS gene
<i>DOCK9</i>	1	1	CMC	5' UTR variants	non-EDS gene
<i>FASLG</i>	1	1	CMC	5' UTR variants	non-EDS gene
<i>TF</i>	1	1	CMC	5' UTR variants	non-EDS gene
<i>DOCK9</i>	1	1	CMC	Intronic variants	non-EDS gene
<i>LOX</i>	1	1	CMC	Intronic variants	non-EDS gene

<i>TF</i>	1	1	CMC	Intronic variants	non-EDS gene
<i>TGFB1</i>	1	1	CMC	Intronic variants	non-EDS gene
<i>PARP1</i>	1	1	CMC	Intronic variants	non-EDS gene
<i>HLA-G</i>	1	1	CMC	Synonymous variant	non-EDS gene
<i>LAMC3</i>	1	1	CMC	Synonymous variant	non-EDS gene
<i>LCA5</i>	1	1	CMC	Synonymous variant	non-EDS gene
<i>RDH13</i>	1	1	CMC	Synonymous variant	non-EDS gene
<i>TF</i>	1	1	CMC	Synonymous variant	non-EDS gene
<i>SOD1</i>	1	1	CMC	Synonymous variant	non-EDS gene
<i>EPPK1</i>	1	1	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>LAMC3</i>	1	1	skat	Nonsynonymous variants and in-frame indels	non-EDS gene
<i>COL2A1</i>	1	1	skat	3' UTR variants	non-EDS gene
<i>NLRP1</i>	1	1	skat	Intronic variants	non-EDS gene

MAF = Minor Allele Frequency; Vt = Variable Thresholds methods; kbac = kernel-based adaptive cluster; CMC = Combined and Multivariate collapsing; skat = SNP-set (Sequence) Kernel Association Method; EDS = Ehlers-Danlos Syndrome. Hg19 was used as reference genome.

**Supplemental table S5.** Identified variants in *COL5A1* with MAF<1% in the control population (according to NM\_000093.4). Reference genome is hg19.

Genomic position	Variant alleles in patients/ Total number of patient alleles	Variant alleles in controls/ Total number of control alleles	cDNA change	Amino Acid change	p-value
chr9:137582774C>T	1/1468	1/1604	c.126C>T	p.(=)	1.0000
chr9:137582841C>T	7/1486	6/1618	c.193C>T	p.(Arg65Trp)	0.7835
chr9:137591755C>T	4/1490	6/1618	c.278C>T	p.(Ala93Val)	0.7556
chr9:137591756G>A	1/1490	2/1618	c.279G>A	p.(=)	1.0000
chr9:137591773A>C	1/1490	0/1618	c.296A>C	p.(Asp99Ala)	0.4794
chr9:137591801C>T	0/1490	2/1618	c.324C>T	p.(=)	0.5007
chr9:137591818C>A	3/1438	0/1582	c.341C>A	p.(Ala114Asp)	0.1078
chr9:137591855G>T	0/1472	1/1606	c.378G>T	p.(Gln126His)	1.0000
chr9:137593039G>T	1/1486	3/1618	c.514G>T	p.(Val172Phe)	0.6261
chr9:137593069G>A	1/1486	0/1618	c.544G>A	p.(Asp182Asn)	0.4787
chr9:137593098C>T	9/1488	0/1618	c.573C>T	p.(=)	0.0013
chr9:137593099G>A	15/1488	14/1618	c.574G>A	p.(Asp192Asn)	0.7121
chr9:137593102C>T	0/1488	1/1618	c.577C>T	p.(Arg193Cys)	1.0000
chr9:137593123G>A	1/1038	1/1074	c.598G>A	p.(Asp200Asn)	1.0000
chr9:137620475G>A	1/1170	0/1368	c.787-41G>A		0.4610
chr9:137620688C>A	1/300	0/492	c.924+35C>A		0.3788
chr9:137622093G>A	1/1286	2/1472	c.936G>A	p.(=)	1.0000
chr9:137622153C>T	3/1432	0/1598	c.996C>T	p.(=)	0.1054
chr9:137622157G>A	0/1432	1/1596	c.1000G>A	p.(Gly334Ser)	1.0000
chr9:137622219C>T	1/1102	1/1294	c.1062C>T	p.(=)	1.0000
chr9:137623302C>T	0/1420	1/1568	c.1165-40C>T		1.0000
chr9:137623378G>A	0/1422	1/1572	c.1201G>A	p.(Glu401Lys)	1.0000
chr9:137623458G>A	1/1190	0/1338	c.1281G>A	p.(=)	0.4707
chr9:137623918T>C	1/1188	0/1366	c.1334T>C	p.(Ile445Thr)	0.4652
chr9:137623967C>T	3/1186	2/1366	c.1383C>T	p.(=)	0.6685
chr9:137630279G>A	1/368	0/466	c.1390-41G>A		0.4412
chr9:137630284G>A	0/368	1/464	c.1390-36G>A		1.0000
chr9:137630288C>T	3/368	1/466	c.1390-32C>T		0.3263
chr9:137630302_137630303del	0/358	1/452	c.1390-18_1390-17del		1.0000
chr9:137630541T>C	0/1036	2/1256	c.1432-51T>C		0.5044
chr9:137630560T>C	1/1034	0/1256	c.1432-32T>C		0.4515
chr9:137642420C>T	1/622	0/776	c.1527C>T	p.(=)	0.4449
chr9:137642459G>A	1/626	1/780	c.1566G>A	p.(=)	1.0000
chr9:137642816C>T	1/924	0/1214	c.1662+88C>T		0.4322
chr9:137642816C>A	2/926	3/1212	c.1662+88C>A		1.0000
chr9:137648616G>T	1/922	0/1010	c.1833G>T	p.(=)	0.4772
chr9:137648681C>T	0/922	1/1008	c.1881+17C>T		1.0000
chr9:137650084T>G	1/1036	1/1242	c.1882-5T>G		1.0000
chr9:137650150T>G	1/1038	1/1242	c.1935+8T>G		1.0000
chr9:137650154C>T	0/1036	1/1240	c.1935+12C>T		1.0000
chr9:137653750G>C	1/1312	2/1536	c.1936-21G>C		1.0000
chr9:137655516C>T	2/1292	3/1478	c.1990-23C>T		1.0000
chr9:137655530C>T	1/1292	0/1478	c.1990-9C>T		0.4664
chr9:137655580G>A	1/1292	1/1478	c.2031G>A	p.(=)	1.0000
chr9:137655607C>T	19/1290	13/1478	c.2034+24C>T		0.1571
chr9:137658291C>T	5/1470	0/1612	c.2089-9C>T		0.0246
chr9:137658303G>C	0/1470	1/1612	c.2092G>C	p.(Val698Leu)	1.0000
chr9:137658304T>A	0/1470	1/1612	c.2093T>A	p.(Val698Asp)	1.0000
chr9:137658307C>T	3/1470	2/1612	c.2096C>T	p.(Thr699Met)	0.6743
chr9:137658877A>G	0/1408	1/1532	c.2165A>G	p.(Gln722Arg)	1.0000
chr9:137659145C>A	1/1198	0/1388	c.2188-11C>A		0.4633
chr9:137659237G>A	1/1198	0/1388	c.2232+37G>A		0.4633
chr9:137659238C>T	0/1200	1/1388	c.2232+38C>T		1.0000
chr9:137660363G>A	1/1138	0/1352	c.2286+54G>A		0.4570
chr9:137666681C>T	2/1450	0/1602	c.2332-24C>T		0.2256
chr9:137666682G>A	0/1448	1/1604	c.2332-23G>A		1.0000
chr9:137666719C>G	1/1450	0/1604	c.2346C>G	p.(=)	0.4748

chr9:137666728G>A	0/1448	1/1604	c.2355G>A	p.(=)	1.0000
chr9:137666787G>A	1/1450	0/1604	c.2385+29G>A		0.4748
chr9:137671999C>T	0/1488	1/1618	c.2430+7C>T		1.0000
chr9:137674521C>T	0/1372	1/1524	c.2439C>T	p.(=)	1.0000
chr9:137674589T>C	0/1368	1/1524	c.2484+23T>C		1.0000
chr9:137676825T>C	0/1248	1/1410	c.2485-10T>C		1.0000
chr9:137676892C>T	1/1250	0/1414	c.2542C>T	p.(Arg848Cys)	0.4692
chr9:137676905A>G	1/1250	1/1414	c.2555A>G	p.(Asn852Ser)	1.0000
chr9:137676914C>G	1/1378	1/1562	c.2564C>G	p.(Pro855Arg)	1.0000
chr9:137687010C>T	0/1490	1/1620	c.2745+38C>T		1.0000
chr9:137687040G>A	0/1490	1/1620	c.2745+68G>A		1.0000
chr9:137688283C>T	1/1126	0/1298	c.2844+19C>T		0.4645
chr9:137688284G>T	0/1126	1/1300	c.2844+20G>T		1.0000
chr9:137688604G>A	0/1418	2/1558	c.2845-90G>A		0.5009
chr9:137688635C>T	12/1418	5/1558	c.2845-59C>T		0.0857
chr9:137690241G>A	0/1490	1/1620	c.2899-13G>A		1.0000
chr9:137694741C>T	0/1438	1/1582	c.3014C>T	p.(Thr1005Met)	1.0000
chr9:137694796C>T	0/1442	1/1584	c.3069C>T	p.(=)	1.0000
chr9:137694814G>A	0/1346	1/1534	c.3087G>A	p.(=)	1.0000
chr9:137694853G>C	0/1488	2/1620	c.3114+12G>C		0.5007
chr9:137694875C>T	1/1488	0/1620	c.3114+34C>T		0.4788
chr9:137694947C>T	1/1488	0/1620	c.3114+106C>T		0.4788
chr9:137696895G>A	1/1410	0/1570	c.3189G>A	p.(=)	0.4732
chr9:137697995G>A	0/830	1/992	c.3259-40G>A		1.0000
chr9:137698067C>T	1/1490	0/1620	c.3291C>T	p.(=)	0.4791
chr9:137698068G>A	1/1490	0/1620	c.3292G>A	p.(Ala1098Thr)	0.4791
chr9:137703169G>C	1/884	0/996	c.3529-16G>C		0.4702
chr9:137703346C>T	0/922	2/1150	c.3591C>T	p.(=)	0.5058
chr9:137703465C>T	1/524	0/696	c.3690+20C>T		0.4295
chr9:137703489C>T	1/742	1/954	c.3690+44C>T		1.0000
chr9:137703495C>A	1/742	1/952	c.3690+50C>A		1.0000
chr9:137704286T>C	1/1488	0/1618	c.3691-9T>C		0.4791
chr9:137704379C>T	2/1490	4/1620	c.3744+31C>T		0.6888
chr9:137704385A>G	0/1490	1/1620	c.3744+37A>G		1.0000
chr9:137704387C>T	0/1490	2/1620	c.3744+39C>T		0.5007
chr9:137704630G>T	1/898	0/1130	c.3852+72G>T		0.4428
chr9:137705831C>T	1/1220	0/1328	c.3855C>T	p.(=)	0.4788
chr9:137705893G>A	1/1214	0/1316	c.3906+11G>A		0.4798
chr9:137705902G>A	0/1212	3/1314	c.3906+20G>A		0.2509
chr9:137705916A>G	1/1218	0/1318	c.3906+34A>G		0.4803
chr9:137706595G>A	1/1146	1/1384	c.3907-48G>A		1.0000
chr9:137706659G>A	1/1138	0/1372	c.3923G>A	p.(Arg1308Lys)	0.4534
chr9:137706772C>T	1/398	0/548	c.4014+22C>T		0.4207
chr9:137707472C>T	0/434	1/642	c.4065C>T	p.(=)	1.0000
chr9:137708884C>T	18/1358	14/1536	c.4135C>T	p.(Pro1379Ser)	0.2926
chr9:137708907G>A	0/1144	1/1310	c.4158G>A	p.(=)	1.0000
chr9:137708915C>T	0/1140	1/1292	c.4166C>T	p.(Pro1389Leu)	1.0000
chr9:137709690G>A	0/372	1/422	c.4230+13G>A		1.0000
chr9:137710561C>T	1/662	1/904	c.4290C>T	p.(=)	1.0000
chr9:137710672G>T	4/482	4/540	c.4339-22G>T		1.0000
chr9:137710699A>T	0/1108	1/1276	c.4344A>T	p.(Glu1448Asp)	1.0000
chr9:137710744C>T	0/704	1/858	c.4389C>T	p.(=)	1.0000
chr9:137710765T>C	1/710	0/870	c.4392+18T>C		0.4494
chr9:137710777C>G	14/1212	6/1426	c.4392+30C>G		0.0408
chr9:137710792C>T	14/1212	6/1426	c.4392+45C>T		0.0408
chr9:137710827C>T	0/622	1/860	c.4393-19C>T		1.0000
chr9:137710829C>T	5/616	4/860	c.4393-17C>T		0.5031
chr9:137710863C>T	3/646	4/890	c.4410C>T	p.(=)	1.0000
chr9:137710938G>A	0/660	1/926	c.4446+39G>A		1.0000
chr9:137712022C>T	0/1480	1/1616	c.4507C>T	p.(Arg1503Cys)	1.0000
chr9:137715276G>A	0/1374	1/1528	c.4659G>A	p.(=)	1.0000
chr9:137715293A>C	0/1306	2/1468	c.4676A>C	p.(His1559Pro)	0.5015
chr9:137715300A>C	0/1304	1/1468	c.4683A>C	p.(=)	1.0000
chr9:137716529C>T	0/1012	2/1302	c.4782C>T	p.(=)	0.5076

chr9:137716542G>A	1/1460	0/1618	c.4795G>A	p.(Glu1599Lys)	0.4743
chr9:137716652C>T	0/1430	1/1604	c.4905C>T	p.(=)	1.0000
chr9:137717676G>A	0/1334	2/1508	c.4993G>A	p.(Asp1665Asn)	0.5017
chr9:137726950C>T	1/308	0/474	c.5270C>T	p.(Thr1757Met)	0.3939
chr9:137727030G>A	0/312	1/552	c.5350G>A	p.(Ala1784Thr)	1.0000
chr9:137734039G>A	0/1094	2/1358	c.5407G>A	p.(Asp1803Asn)	0.5056
chr9:137734101G>A	1/1490	0/1620	c.5469G>A	p.(=)	0.4791
chr9:137734101G>C	1/1490	0/1620	c.5469G>C	p.(=)	0.4791
Sum over all variant positions	207/150270	183/171234			

MAF = Minor Allele Frequency; chr = chromosome.

**Supplemental table S6.** Identified variants in *COL2A1* with MAF<0.1% in the control population (according to NM\_001844.4). Hg19 was used as reference genome.

Genomic position	Variant alleles in patients/ Total number of patient alleles	Variant alleles in controls/ Total number of control alleles	cDNA change	Amino Acid change	p-value
chr12:48367305A>G	1/1292	0/1500	c.4349T>C	p.(Ile1450Thr)	0.4628
chr12:48367771T>G	1/1088	0/1288	c.4317+101A>C		0.4579
chr12:48368073G>A	1/1426	0/1598	c.4116C>T	p.(=)	0.4716
chr12:48368174G>T	0/1490	1/1620	c.4075-60C>A		1.0000
chr12:48369159C>T	1/1488	0/1620	c.3827G>A	p.(Arg1276His)	0.4788
chr12:48369250C>T	1/1442	1/1596	c.3736G>A	p.(Gly1246Ser)	1.0000
chr12:48369262C>T	0/1442	1/1596	c.3724G>A	p.(Asp1242Asn)	1.0000
chr12:48369263G>A	1/1442	0/1596	c.3723C>T	p.(=)	0.4747
chr12:48369287G>A	1/828	0/1046	c.3699C>T	p.(=)	0.4418
chr12:48369832G>A	1/1450	0/1588	c.3511C>T	p.(Pro1171Ser)	0.4773
chr12:48369872G>T	1/1444	0/1582	c.3490-19C>A		0.4772
chr12:48370870C>T	0/1286	1/1386	c.3327+15G>A		1.0000
chr12:48370896G>A	1/1286	0/1386	c.3316C>T	p.(Arg1106Trp)	0.4813
chr12:48371063C>T	1/826	0/910	c.3273+40G>A		0.4758
chr12:48371075C>T	2/826	0/916	c.3273+28G>A		0.2247
chr12:48374400G>A	1/400	0/616	c.2562C>T	p.(=)	0.3937
chr12:48375548C>T	1/206	0/420	c.2409+11G>A		0.3291
chr12:48376305C>T	1/448	0/626	c.2281G>A	p.(Ala761Thr)	0.4171
chr12:48376864C>G	4/1190	1/1426	c.2094+12G>C		0.1839
chr12:48376864C>T	1/1188	0/1424	c.2094+12G>A		0.4548
chr12:48376876C>G	0/1190	1/1426	c.2094G>C	p.(Arg698Ser)	1.0000
chr12:48377957C>T	3/1168	1/1402	c.1888-34G>A		0.3357
chr12:48379494G>A	2/1228	0/1358	c.1680+17C>T		0.2254
chr12:48379502G>T	2/1228	0/1356	c.1680+9C>A		0.2257
chr12:48379503C>A	2/1228	0/1356	c.1680+8G>T		0.2257
chr12:48379557T>C	0/1234	1/1358	c.1634A>G	p.(Asn545Ser)	1.0000
chr12:48379675G>C	1/848	0/896	c.1581+20C>G		0.4862
chr12:48379722T>C	0/1380	1/1546	c.1554A>G	p.(=)	1.0000
chr12:48379804G>A	1/1268	0/1442	c.1528-56C>T		0.4679
chr12:48380239A>C	1/854	0/1064	c.1420-13T>G		0.4453
chr12:48380908G>T	1/464	0/644	c.1318C>A	p.(Pro440Thr)	0.4188
chr12:48381033del	1/990	0/1238	c.1266+19del		0.4443
chr12:48381439G>A	2/1476	1/1616	c.1176C>T	p.(=)	0.6087
chr12:48383006C>T	1/1374	0/1486	c.1122+5G>A		0.4804
chr12:48383056G>C	0/1374	1/1486	c.1077C>G	p.(=)	1.0000
chr12:48386666C>T	0/1490	1/1620	c.1018G>A	p.(Ala340Thr)	1.0000
chr12:48386743A>G	0/1490	1/1620	c.970-29T>C		1.0000
chr12:48387211T>C	1/1490	0/1620	c.969+30A>G		0.4791
chr12:48387243T>C	0/1490	1/1620	c.967A>G	p.(Met323Val)	1.0000
chr12:48387250C>T	0/1490	1/1620	c.960G>A	p.(=)	1.0000
chr12:48387294A>G	1/1490	0/1620	c.925-9T>C		0.4791
chr12:48387691A>G	1/1490	0/1620	c.871-46T>C		0.4791
chr12:48387708G>A	1/1480	0/1620	c.871-63C>T		0.4774
chr12:48387755C>A	2/1478	0/1620	c.870+22G>T		0.2275
chr12:48388236C>T	0/1424	1/1534	c.787G>A	p.(Ala263Thr)	1.0000
chr12:48389107G>A	0/1490	1/1616	c.709-16C>T		1.0000
chr12:48389461C>T	0/1490	1/1620	c.708+32G>A		1.0000
chr12:48389485G>A	1/1490	1/1620	c.708+8C>T		1.0000
chr12:48389710G>A	1/1474	0/1590	c.610-8C>T		0.4811
chr12:48389717_48389718del	0/1474	1/1588	c.610-16_610-15del		1.0000
chr12:48389736A>G	1/1474	0/1590	c.610-34T>C		0.4811
chr12:48390320T>C	1/1490	0/1602	c.609+11A>G		0.4819
chr12:48390376A>G	1/1490	0/1620	c.564T>C	p.(=)	0.4791
chr12:48390428C>T	1/1488	0/1620	c.532-20G>A		0.4788
chr12:48391769C>A	0/1470	1/1582	c.375+46G>T		1.0000
chr12:48391788G>A	0/1476	1/1586	c.375+27C>T		1.0000
chr12:48391929G>A	1/1490	1/1620	c.342+23C>T		1.0000

chr12:48392004C>T	0/1490	1/1620	c.310-20G>A		1.0000
chr12:48392015C>T	1/1490	0/1620	c.310-31G>A		0.4791
chr12:48393783C>T	1/1486	0/1594	c.211G>A	p.(Asp71Asn)	0.4825
chr12:48398075C>G	0/1486	1/1612	c.30G>C	p.(=)	1.0000
Sum over all variant positions	53/78352	25/87228			

MAF = Minor Allele Frequency; chr = chromosome.



**Supplemental table S7.** Identified nonsynonymous variants in *TNXB* with MAF<0.1% in the control population (according to NM\_019105.6). Hg19 was used as reference genome.

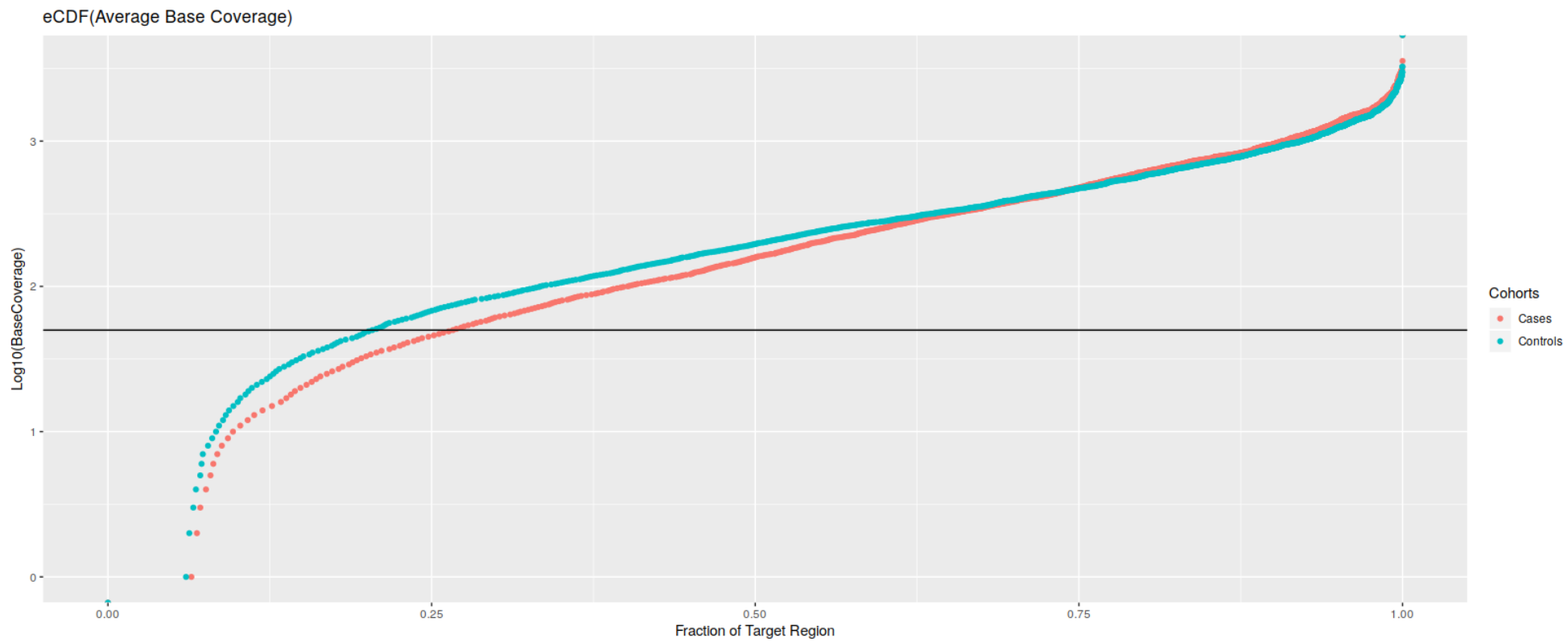
Genomic position	Variant alleles in patients/ Total number of patient alleles	Variant alleles in controls/ Total number of control alleles	cDNA change	Amino Acid change	p-value
chr6:32013984C>T	0/1260	1/1454	c.10568G>A	p.(Arg3523His)	1.0000
chr6:32014069C>T	2/562	0/694	c.10483G>A	p.(Ala3495Thr)	0.2000
chr6:32016167C>T	1/858	0/952	c.10012G>A	p.(Gly3338Ser)	0.4740
chr6:32020479C>T	2/1448	0/1596	c.9077G>A	p.(Arg3026His)	0.2262
chr6:32023663C>T	1/932	0/1040	c.8432G>A	p.(Arg2811Gln)	0.4726
chr6:32035489C>T	0/1062	1/1322	c.6493G>A	p.(Gly2165Ser)	1.0000
chr6:32036313T>A	2/1452	0/1594	c.6074A>T	p.(Asp2025Val)	0.2272
chr6:32036733T>C	0/1466	1/1604	c.5768A>G	p.(Gln1923Arg)	1.0000
chr6:32037978C>T	1/1490	0/1620	c.5204G>A	p.(Arg1735His)	0.4791
chr6:32046842C>T	0/1310	1/1468	c.4343G>A	p.(Arg1448His)	1.0000
chr6:32047080A>C	1/1468	0/1614	c.4105T>G	p.(Ser1369Ala)	0.4763
chr6:32047085G>A	1/1470	0/1614	c.4100C>T	p.(Pro1367Leu)	0.4767
chr6:32049271G>T	0/1418	1/1576	c.3916C>A	p.(Pro1306Thr)	1.0000
chr6:32050061C>T	6/1488	0/1616	c.3488G>A	p.(Gly1163Glu)	0.0121
chr6:32052255T>C	2/1488	0/1618	c.3380A>G	p.(Lys1127Arg)	0.2294
chr6:32057048G>C	0/1438	1/1580	c.2467C>G	p.(Leu823Val)	1.0000
chr6:32063547G>A	1/1456	0/1608	c.2083C>T	p.(Arg695Trp)	0.4752
chr6:32063793T>C	2/1392	0/1520	c.1837A>G	p.(Ser613Gly)	0.2284
chr6:32063858G>A	0/1178	1/1340	c.1772C>T	p.(Pro591Leu)	1.0000
chr6:32063891T>C	1/1478	0/1612	c.1739A>G	p.(Tyr580Cys)	0.4783
chr6:32065011C>T	3/1466	0/1606	c.619G>A	p.(Gly207Ser)	0.1086
Sum over all variant positions	26/27580	7/30648			

MAF = Minor Allele Frequency; chr = chromosome.

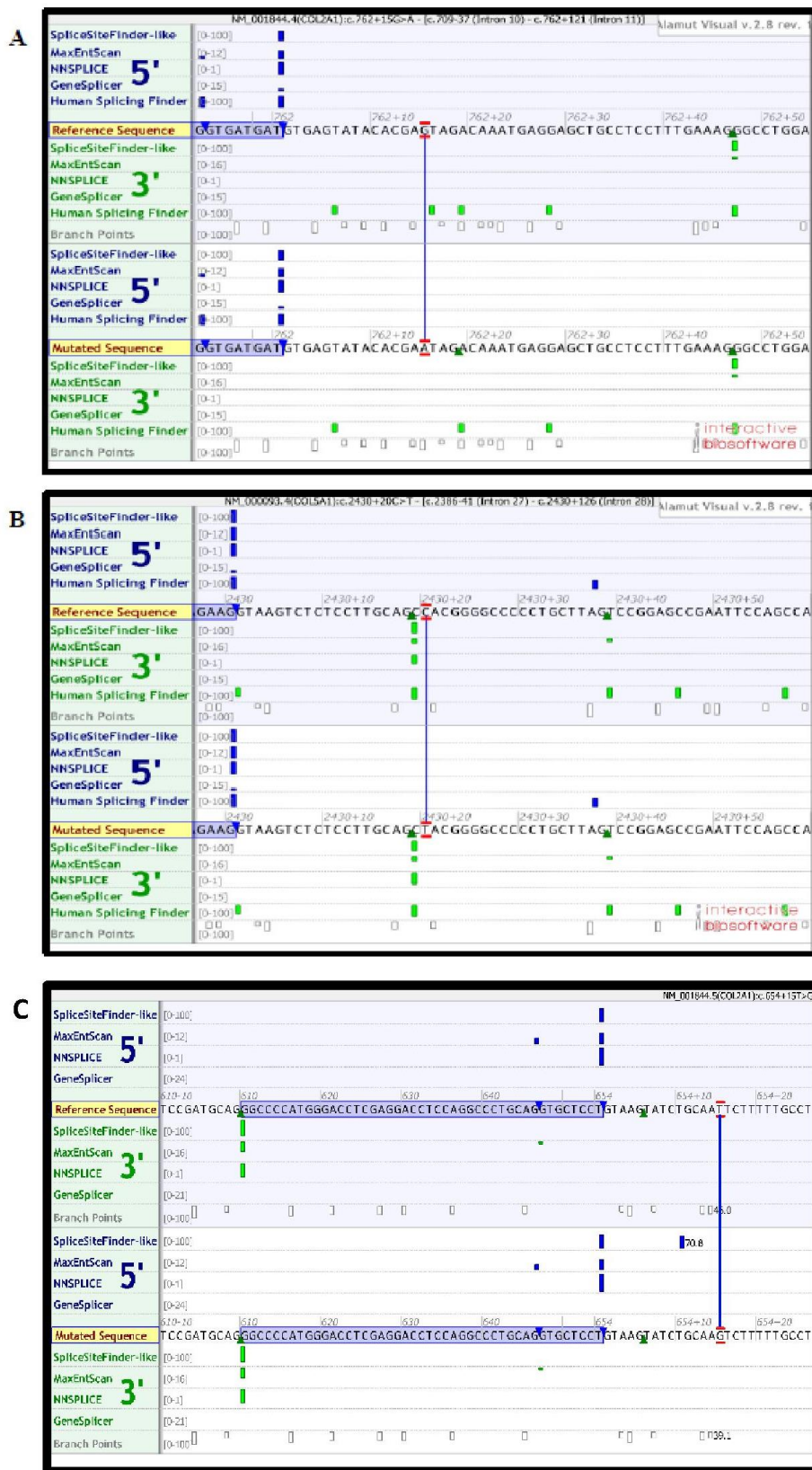
**Supplemental table S8.** Identified variants in *ZNF469* with MAF<0.1% in the control population (according to NM\_001127464.1). Reference genome is hg19.

Genomic position	Variant alleles in patients/ Total number of patient alleles	Variant alleles in controls/ Total number of control alleles	cDNA change	Amino Acid change	p-value
chr16:88494303T>A	1/438	0/630	c.425T>A	p.(Leu142Gln)	0.4101
chr16:88494638G>A	1/712	0/822	c.760G>A	p.(Glu254Lys)	0.4641
chr16:88495425G>C	0/1040	1/1106	c.1547G>C	p.(Gly516Ala)	1.0000
chr16:88495426C>T	1/1040	0/1106	c.1548C>T	p.(=)	0.4846
chr16:88495462G>A	1/1030	0/1098	c.1584G>A	p.(=)	0.4840
chr16:88496440G>A	1/566	0/734	c.2562G>A	p.(=)	0.4354
chr16:88496531C>G	1/422	0/626	c.2653C>G	p.(Leu885Val)	0.4027
chr16:88498105G>A	2/574	0/738	c.4143G>A	p.(=)	0.1912
chr16:88498452G>A	1/1048	0/1134	c.4490G>A	p.(Ser1497Asn)	0.4803
chr16:88498459G>A	1/1048	0/1134	c.4497G>A	p.(=)	0.4803
chr16:88498965C>T	1/1004	0/1142	c.5003C>T	p.(Pro1668Leu)	0.4678
chr16:88499040G>T	3/912	0/1028	c.5078G>T	p.(Cys1693Phe)	0.1037
chr16:88499378C>A	0/1354	1/1506	c.5416C>A	p.(His1806Asn)	1.0000
chr16:88499416G>A	0/1338	1/1484	c.5454G>A	p.(=)	1.0000
chr16:88499426C>A	4/1340	0/1486	c.5464C>A	p.(Pro1822Thr)	0.0504
chr16:88499455G>A	0/1362	1/1516	c.5493G>A	p.(=)	1.0000
chr16:88499910G>A	2/1262	0/1380	c.5948G>A	p.(Gly1983Asp)	0.2281
chr16:88499913C>T	1/1260	1/1380	c.5951C>T	p.(Thr1984Met)	1.0000
chr16:88500928T>C	1/1066	0/1186	c.6966T>C	p.(=)	0.4734
chr16:88500958G>A	0/1066	1/1186	c.6996G>A	p.(=)	1.0000
chr16:88502084G>A	1/592	0/728	c.8122G>A	p.(Asp2708Asn)	0.4485
chr16:88502477G>A	0/896	1/1042	c.8515G>A	p.(Gly2839Ser)	1.0000
chr16:88502520G>T	0/892	1/1038	c.8558G>T	p.(Arg2853Leu)	1.0000
chr16:88502746C>T	1/966	0/1106	c.8784C>T	p.(=)	0.4662
chr16:88502957C>T	1/422	0/618	c.8995C>T	p.(=)	0.4058
chr16:88503194C>T	0/1000	1/1162	c.9232C>T	p.(Arg3078Trp)	1.0000
chr16:88503197C>T	1/998	0/1160	c.9235C>T	p.(Pro3079Ser)	0.4625
chr16:88503199G>A	1/1000	0/1162	c.9237G>A	p.(=)	0.4625
chr16:88503477G>A	2/364	0/526	c.9515G>A	p.(Arg3172His)	0.1670
chr16:88503869G>T	1/538	0/814	c.9907G>T	p.(Ala3303Ser)	0.3979
chr16:88504396G>A	4/762	0/830	c.10434G>A	p.(=)	0.0523
chr16:88504421G>A	1/472	0/638	c.10459G>A	p.(Val3487Met)	0.4252
chr16:88504477C>T	2/896	0/1000	c.10515C>T	p.(=)	0.2232
chr16:88504534G>A	2/904	1/1018	c.10572G>A	p.(=)	0.6039
chr16:88504536C>T	2/906	0/1018	c.10574C>T	p.(Pro3525Leu)	0.2216
chr16:88504586G>A	2/1064	0/1202	c.10624G>A	p.(Ala3542Thr)	0.2204
chr16:88504604G>C	1/1064	0/1204	c.10642G>C	p.(Glu3548Gln)	0.4691
chr16:88504686G>A	2/1402	0/1584	c.10724G>A	p.(Arg3575Gln)	0.2204
chr16:88504733G>A	0/1364	1/1526	c.10771G>A	p.(Val3591Met)	1.0000
chr16:88504733G>C	0/1364	1/1526	c.10771G>C	p.(Val3591Leu)	1.0000
chr16:88504858G>A	1/1210	0/1322	c.10896G>A	p.(=)	0.4779
chr16:88504985G>T	1/536	0/684	c.11023G>T	p.(Val3675Leu)	0.4393
chr16:88505256C>T	1/1054	0/1170	c.11294C>T	p.(Pro3765Leu)	0.4739
chr16:88505444C>T	1/422	0/588	c.11482C>T	p.(Arg3828Cys)	0.4178
Sum over all variant positions	50/40970	12/47088			

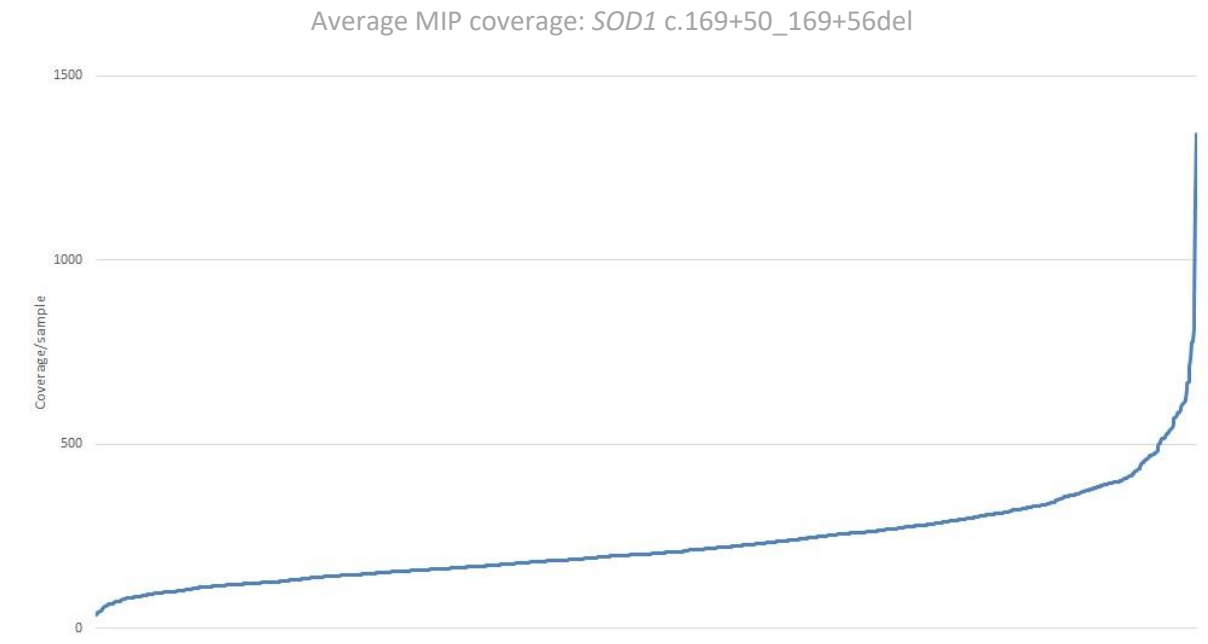
MAF = Minor Allele Frequency; chr = chromosome.



**Supplemental figure S1.** Average base coverage for the target region for case and control samples.

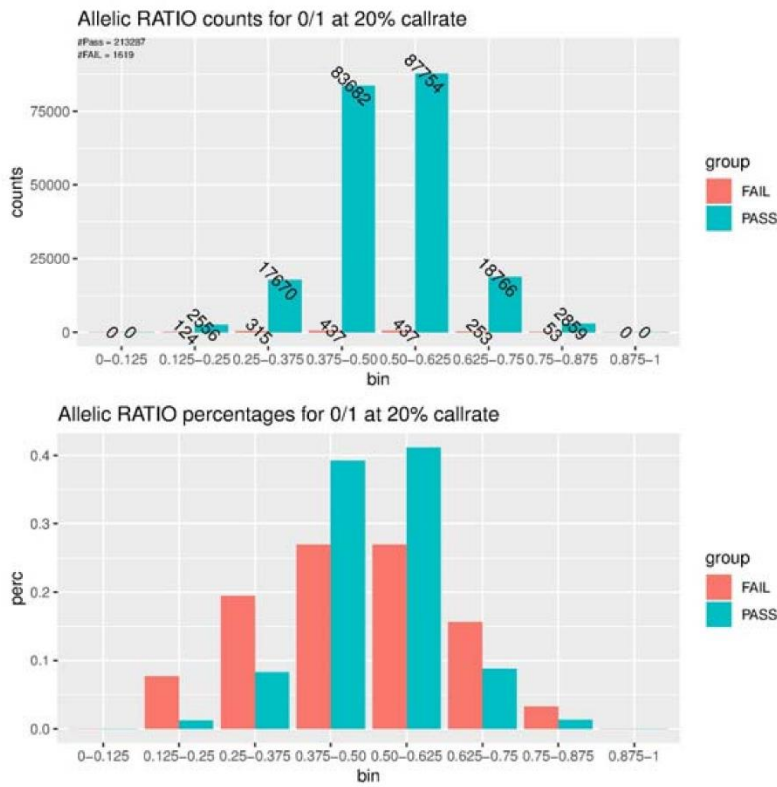


**Supplemental figure S2.** Predictions of the effect on splicing of the three variants located in the exon-intron boundaries and ranking in the top-15 of single SNP association tests.

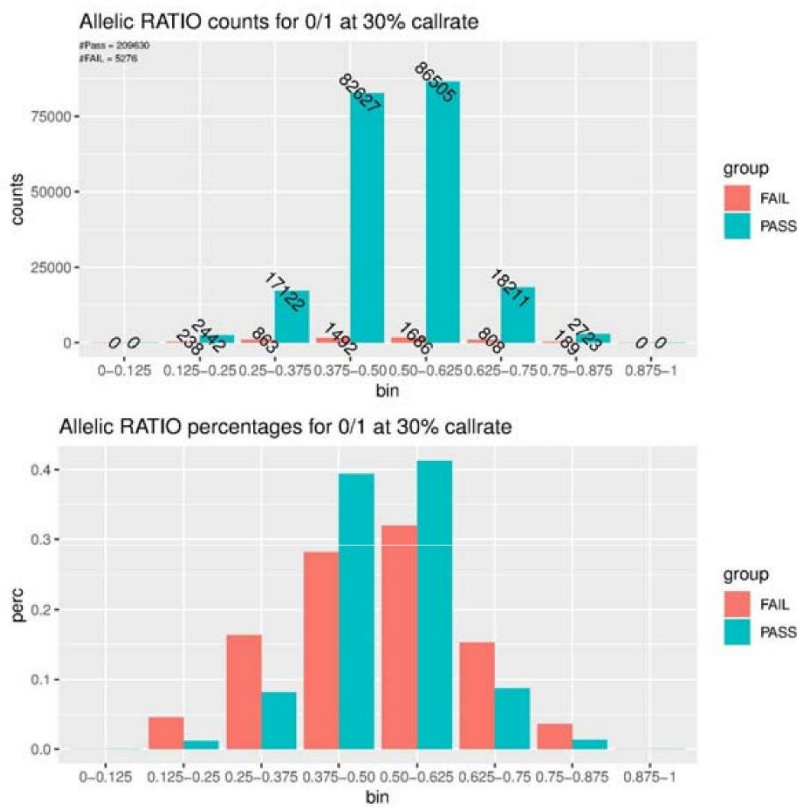


**Supplemental figure S3.** Coverage of the MIP probe targeting the reported *SOD1* c.169+50\_169+56del variant for all included samples. Hg19 was used as reference genome (NC\_000021.8).

**A**



**B**



**Supplemental figure S4.** Histograms of variants passing and failing the criterion.

# Average Coverage Overview

## Transcript summary

Gene	Transcript	Size	Median.Cov	Avg.%@50X	95%CI(%@50X)
COL12A1	NM_004370.5	10,492bp	531.7X	89.7%	(89.5%,90%)
COL12A1	NM_080645.2	6,700bp	495X	89.5%	(89.3%,89.8%)
COL2A1	NM_001844.4	5,544bp	82X	65.2%	(64.4%,66%)
COL2A1	NM_033150.2	5,317bp	82X	65.7%	(64.9%,66.5%)
COL5A1	NM_000093.4	6,837bp	88.5X	70.2%	(69.4%,71.1%)
COL5A1	NM_001278074.1	6,837bp	86X	69.2%	(68.4%,70.1%)
COL5A3	NM_015719.3	6,578bp	107X	70%	(69.4%,70.6%)
DCAF11	NM_001163484.1	1,921bp	374.5X	95.9%	(95.6%,96.1%)
DCAF11	NM_025230.4	1,921bp	374.5X	95.9%	(95.6%,96.1%)
DCAF11	NM_181357.2	1,843bp	391X	95.7%	(95.5%,96%)
DOCK1	NM_001290223.1	6,701bp	699X	93.7%	(93.5%,93.8%)
DOCK1	NM_001380.4	6,638bp	706X	94.5%	(94.4%,94.7%)
DOCK9	NM_001130048.1	7,347bp	561X	93%	(92.8%,93.2%)
DOCK9	NM_001130049.1	4,425bp	644.5X	91%	(90.9%,91.2%)
DOCK9	NM_001130050.1	4,422bp	655.5X	93.7%	(93.6%,93.9%)
DOCK9	NM_001318849.1	7,261bp	559X	91.3%	(91.1%,91.5%)
DOCK9	NM_015296.2	7,350bp	551.5X	91.4%	(91.2%,91.6%)
EPPK1	NM_031308.3	7,300bp	44X	49.4%	(48.1%,50.7%)
FASLG	NM_000639.2	926bp	313X	97.2%	(96.8%,97.6%)
FASLG	NM_001302746.1	424bp	170X	95.4%	(94.7%,96%)
FNDC3B	NM_001135095.1	4,115bp	717X	95.4%	(95.2%,95.6%)
FNDC3B	NM_022763.3	4,115bp	717X	95.4%	(95.2%,95.6%)
FOXO1	NM_002015.3	2,008bp	143X	66.2%	(65.8%,66.6%)
HLA-G	NM_002127.5	1,137bp	120X	68.8%	(68.2%,69.4%)
IL1RN	NM_000577.4	580bp	545X	99.2%	(99.1%,99.4%)
IL1RN	NM_001318914.1	512bp	551X	99.5%	(99.4%,99.6%)
IL1RN	NM_173841.2	663bp	493X	99.3%	(99.2%,99.5%)
IL1RN	NM_173842.2	614bp	496X	99%	(98.8%,99.2%)
IL1RN	NM_173843.2	512bp	551X	99.5%	(99.4%,99.6%)
LAMC3	NM_006059.3	5,288bp	172X	79.6%	(79.1%,80.1%)
LCA5	NM_001122769.2	2,234bp	570.5X	96.4%	(96.2%,96.6%)
LCA5	NM_181714.3	2,234bp	570.5X	96.4%	(96.2%,96.6%)
LOX	NM_001178102.2	684bp	586X	93.7%	(93.2%,94.1%)
LOX	NM_001317073.1	443bp	569X	94.1%	(93.6%,94.5%)
LOX	NM_002317.6	1,394bp	304X	92.6%	(92.1%,93.1%)
LOXHD1	NM_001145472.2	3,785bp	388X	86.2%	(86%,86.4%)
LOXHD1	NM_001145473.2	1,699bp	243X	76.4%	(76%,76.7%)
LOXHD1	NM_001173129.1	1,554bp	299X	79.1%	(78.8%,79.4%)
LOXHD1	NM_001308013.1	3,416bp	396X	87.4%	(87.2%,87.6%)
LOXHD1	NM_144612.6	7,436bp	376X	88.6%	(88.4%,88.9%)
MIR184.nc	NR_029705.1	104bp	203.2X	91.7%	(90.8%,92.5%)
MMP9	NM_004994.2	2,384bp	39X	46.2%	(45.1%,47.2%)
NLRP1	NM_001033053.2	4,448bp	445.5X	91.4%	(91.1%,91.7%)
NLRP1	NM_014922.4	4,610bp	451X	93%	(92.7%,93.3%)
NLRP1	NM_033004.3	4,762bp	449X	91.9%	(91.6%,92.3%)
NLRP1	NM_033006.3	4,652bp	445X	91.8%	(91.5%,92.2%)
NLRP1	NM_033007.3	4,500bp	446X	92.9%	(92.6%,93.3%)
P3H4	NM_006455.2	1,474bp	50X	55%	(54%,55.9%)
PARP1	NM_001618.3	3,505bp	411X	92.8%	(92.5%,93%)
PARP2	NM_001042618.1	2,033bp	604X	89.1%	(88.9%,89.4%)
PARP2	NM_005484.3	2,072bp	617.5X	89.3%	(89.1%,89.6%)
PRPH2	NM_000322.4	1,101bp	372X	97.9%	(97.6%,98.2%)
RDH13	NM_001145971.1	1,136bp	153.7X	85%	(84.4%,85.6%)
RDH13	NM_138412.3	883bp	164.5X	83.9%	(83.3%,84.5%)
SLC4A11	NM_001174089.1	3,028bp	43X	48.8%	(47.5%,50.1%)
SLC4A11	NM_001174090.1	3,157bp	44X	49.1%	(47.8%,50.4%)
SLC4A11	NM_032034.3	3,056bp	42X	48.3%	(47%,49.6%)
SOD1	NM_000454.4	565bp	250X	89.2%	(88.9%,89.5%)
TF	NM_001063.3	2,437bp	396.5X	93%	(92.7%,93.4%)
TGFBI	NM_000358.2	2,392bp	361.7X	90.7%	(90.3%,91.2%)

Continued on next page

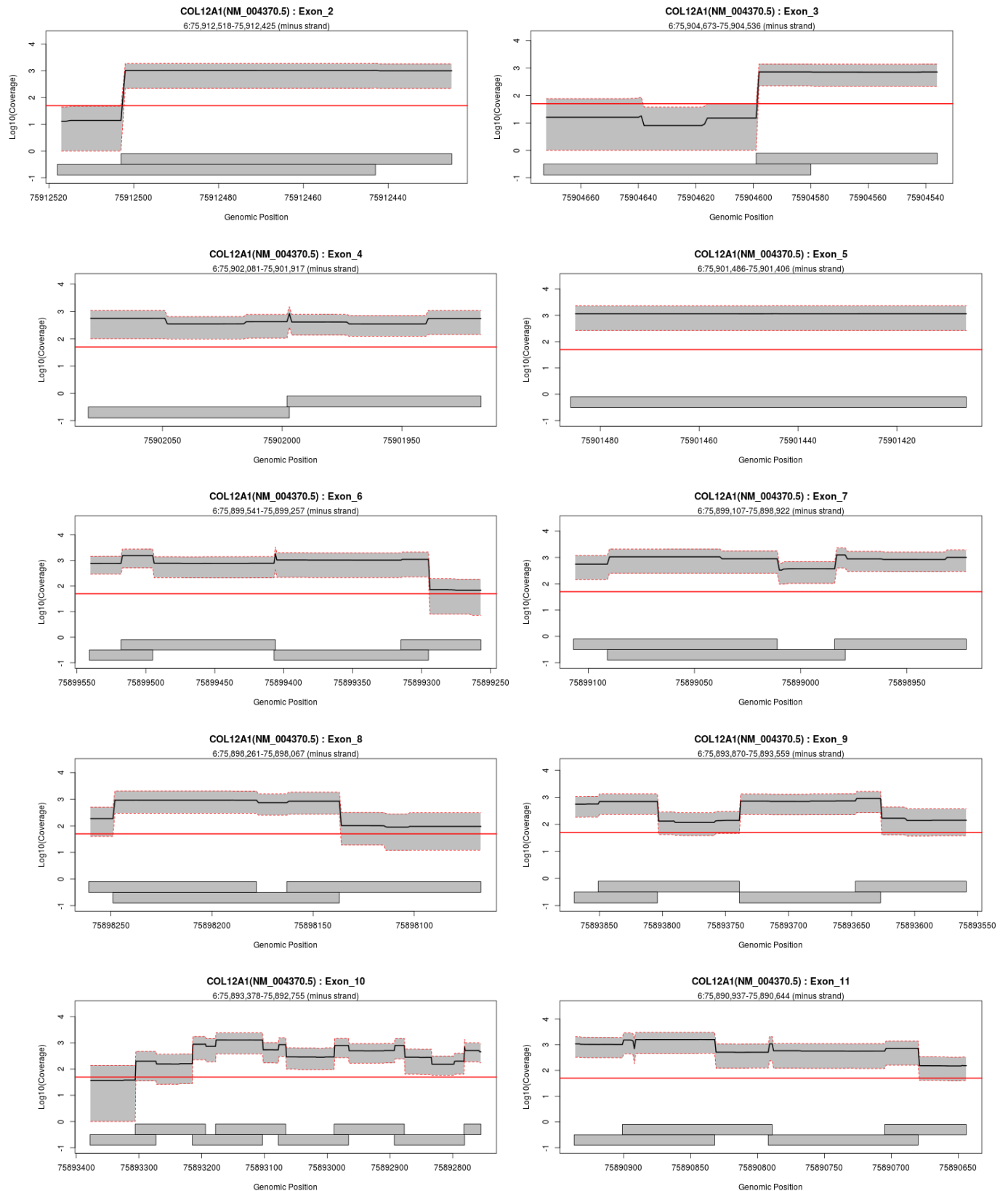
Gene	Transcript	Size	Median.Cov	Avg.%@50X	95%CI(%@50X)
TNXB	NM_019105.6	13,589bp	48X	49.8%	(49%,50.6%)
TNXB	NM_032470.3	2,282bp	11X	31.6%	(30.8%,32.3%)
VSX1	NM_001256271.1	791bp	79X	60.9%	(60.2%,61.6%)
VSX1	NM_001256272.1	1,006bp	96X	67%	(66.3%,67.7%)
VSX1	NM_014588.5	1,198bp	108X	69.4%	(68.8%,70%)
VSX1	NM_199425.2	780bp	103X	60.9%	(60.3%,61.6%)
WNT10A	NM_025216.2	1,334bp	22X	29.9%	(28.8%,31.1%)
ZEB1	NM_001128128.2	3,507bp	445X	93.8%	(93.5%,94%)
ZEB1	NM_001174093.1	3,475bp	448X	91.5%	(91.3%,91.8%)
ZEB1	NM_001174094.1	3,504bp	445X	93.8%	(93.5%,94%)
ZEB1	NM_001174095.1	3,334bp	432X	91.4%	(91.1%,91.6%)
ZEB1	NM_001174096.1	3,558bp	437X	91.7%	(91.4%,91.9%)
ZEB1	NM_001323638.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323641.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323642.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323643.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323644.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323645.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323646.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323647.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323648.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323649.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323650.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323651.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323652.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323653.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323654.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323655.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323656.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323657.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323658.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323659.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323660.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323661.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323662.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323663.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323664.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323665.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323666.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323671.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323672.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323673.1	2,821bp	436X	93%	(92.7%,93.2%)
ZEB1	NM_001323674.1	3,293bp	424X	91.1%	(90.8%,91.3%)
ZEB1	NM_001323675.1	3,251bp	430X	92.3%	(92%,92.5%)
ZEB1	NM_001323676.1	3,516bp	442.5X	92.8%	(92.5%,93%)
ZEB1	NM_001323677.1	3,513bp	443X	92.8%	(92.5%,93%)
ZEB1	NM_001323678.1	3,242bp	431X	93.3%	(93.1%,93.6%)
ZEB1	NM_030751.5	3,555bp	437X	91.7%	(91.4%,91.9%)
ZNF469	NM_001127464	11,902bp	18X	29.8%	(28.5%,31.2%)

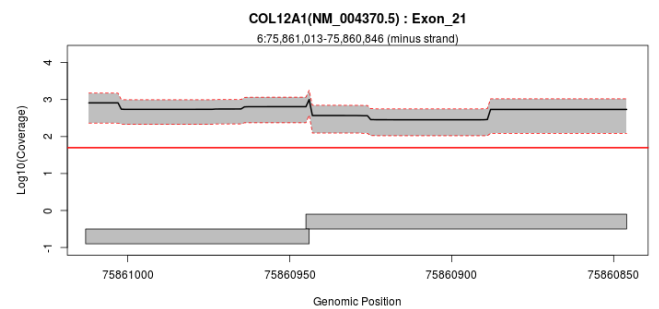
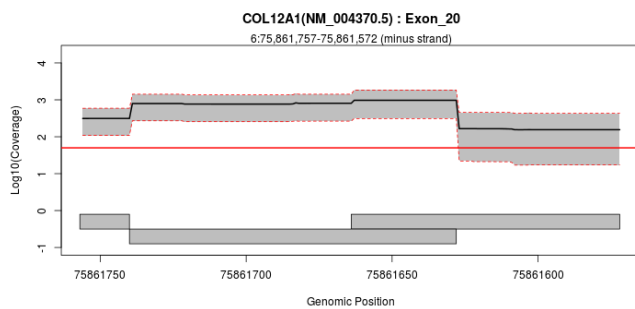
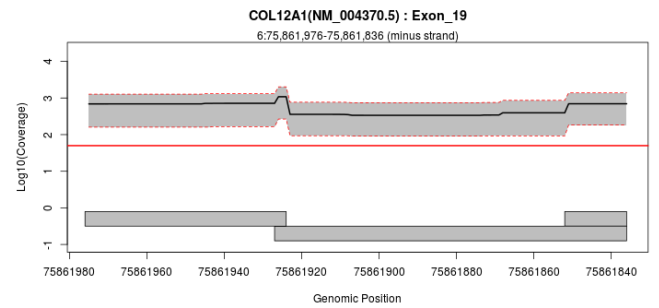
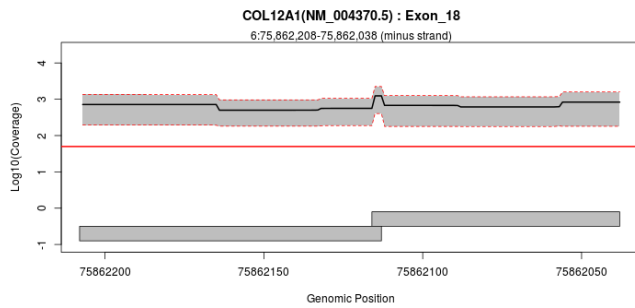
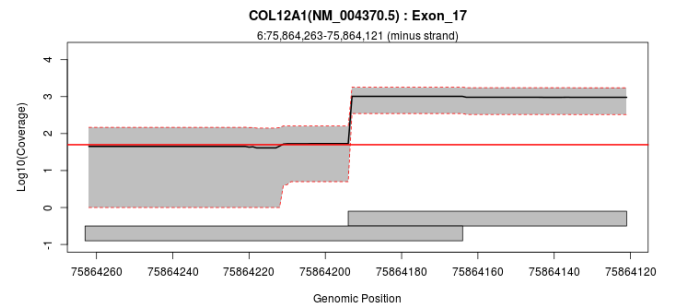
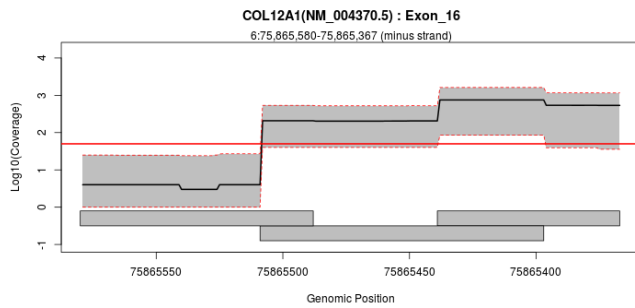
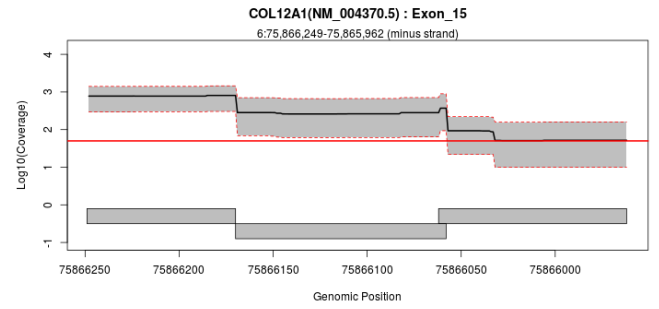
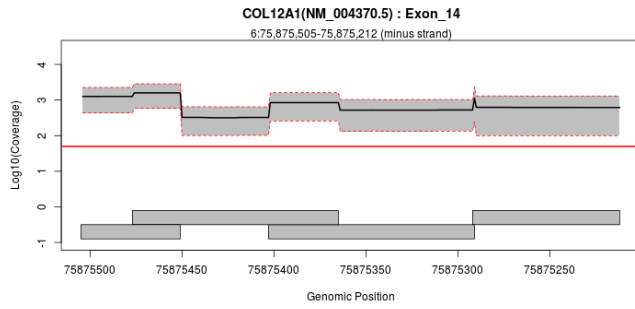
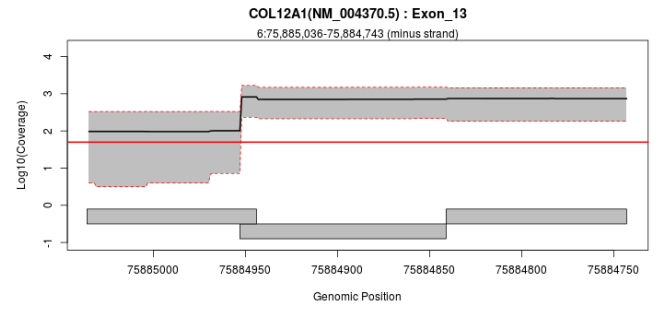
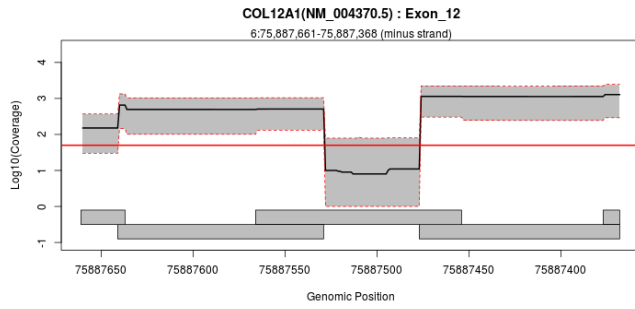


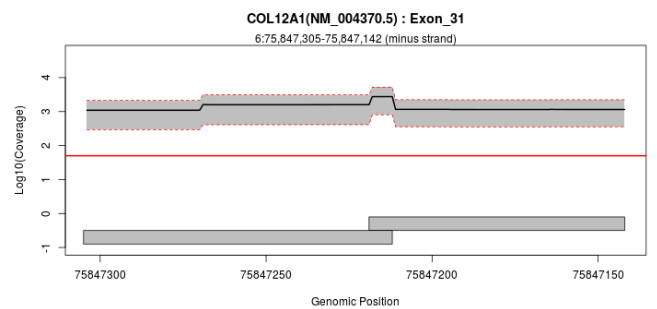
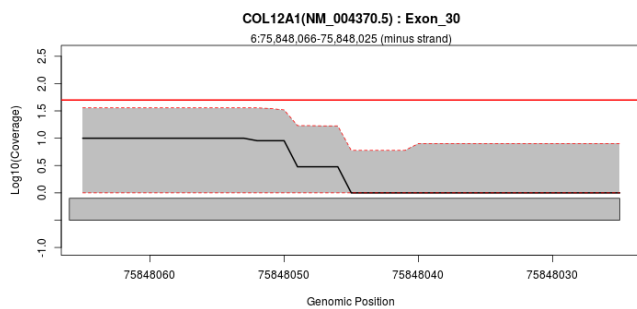
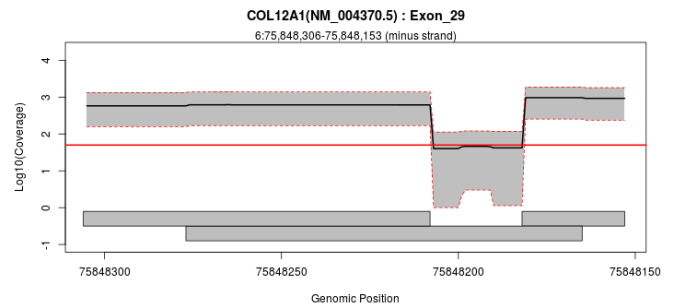
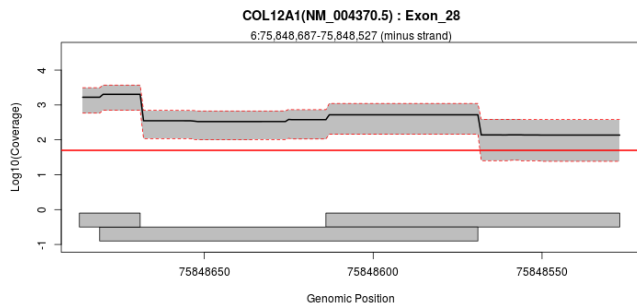
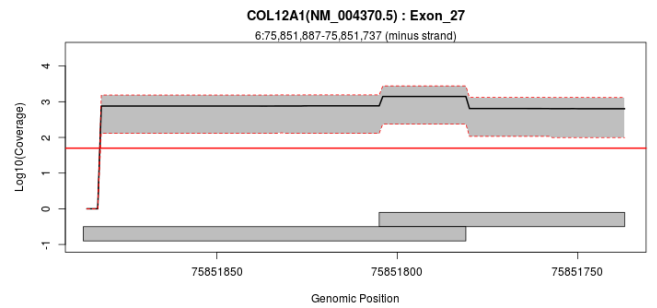
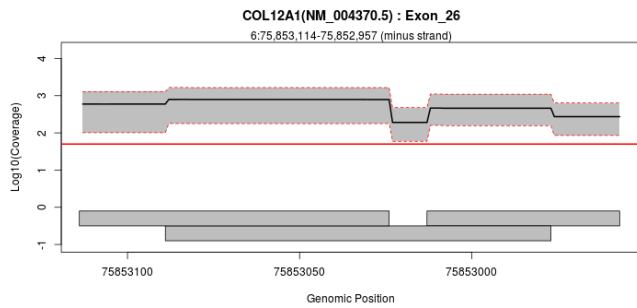
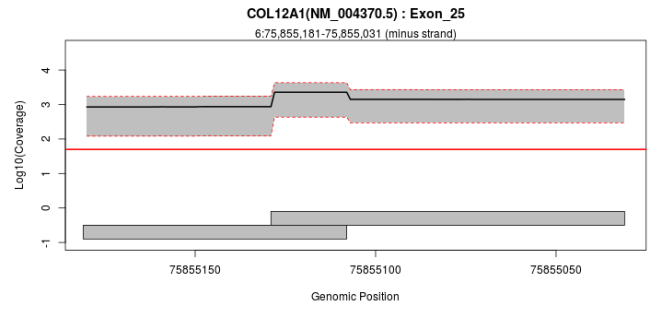
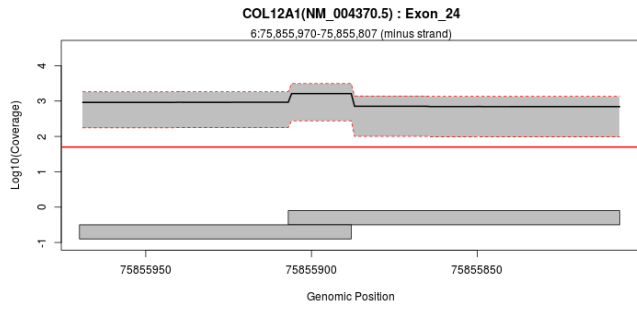
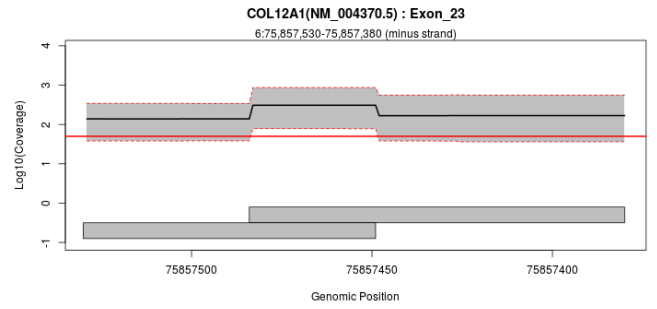
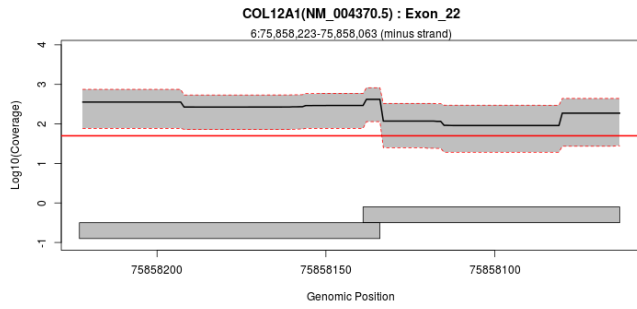
# Detailed Coverage Plots

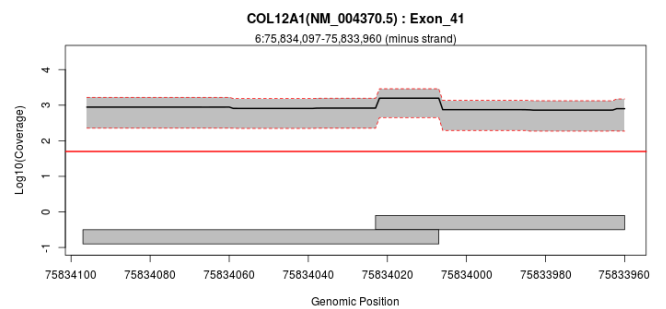
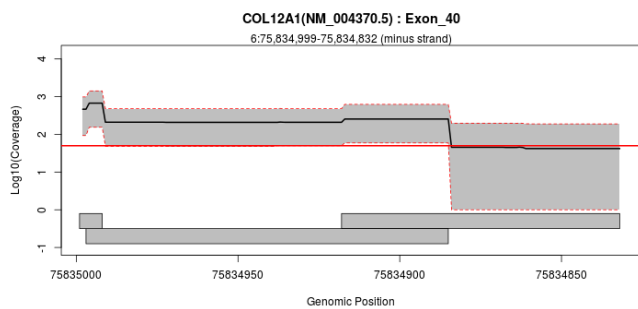
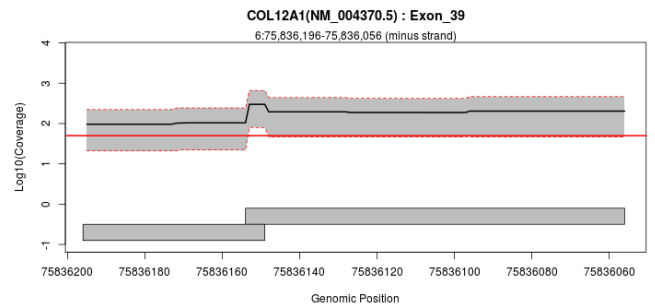
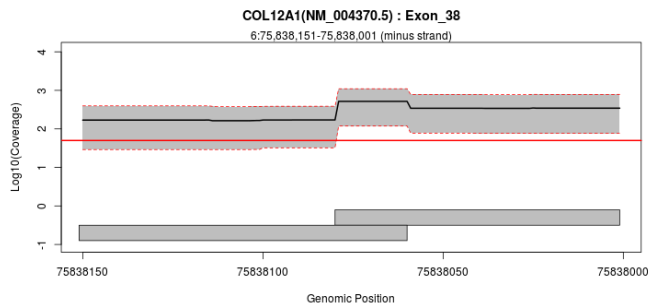
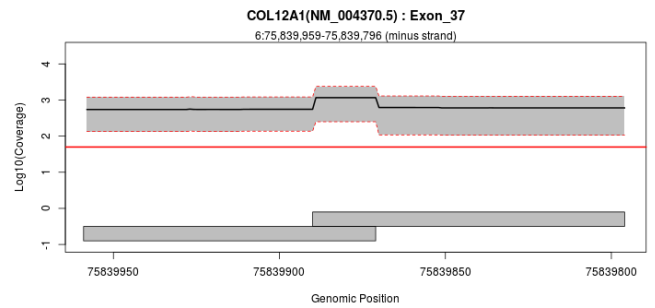
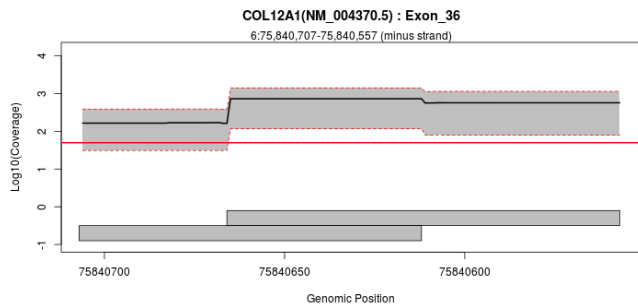
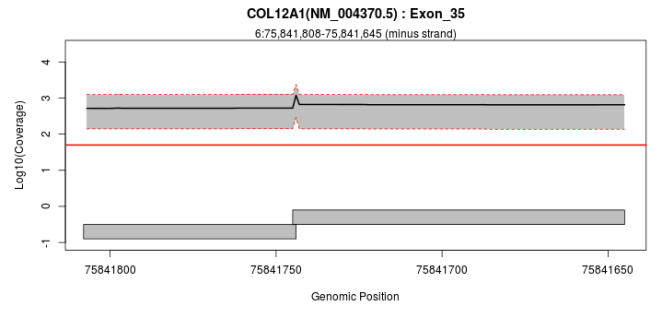
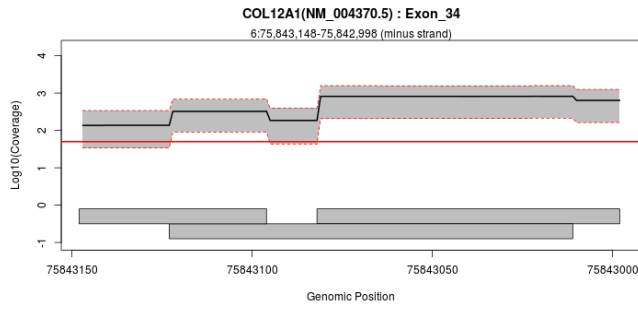
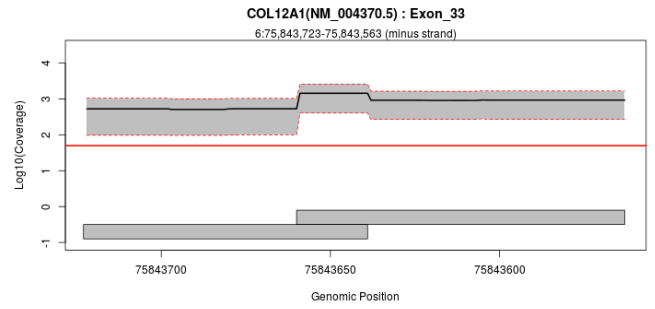
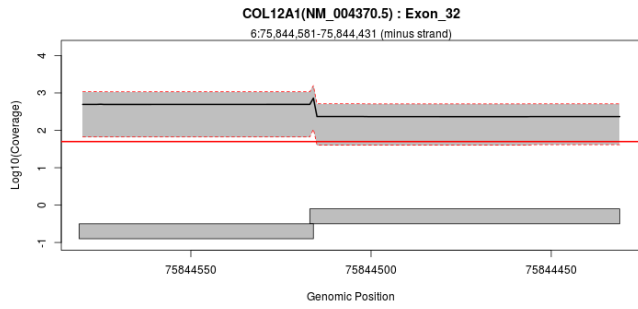
NOTE: In the Coverage plots below, the confidence intervals correspond to 2.5% and 97.5% Quantiles. The black line corresponds to the population median. The red line represents the 50x coverage target. Grey bars represent the MIPS used to capture the region.

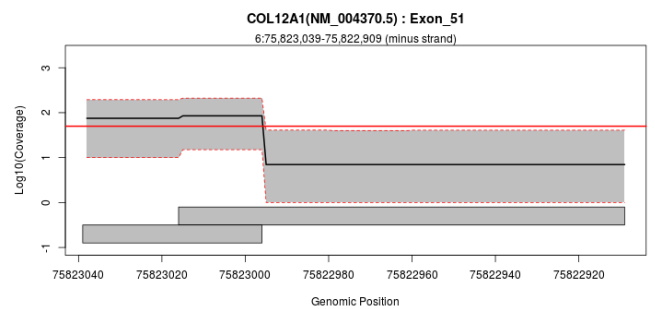
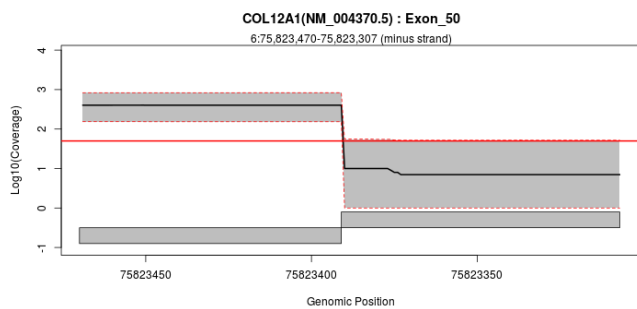
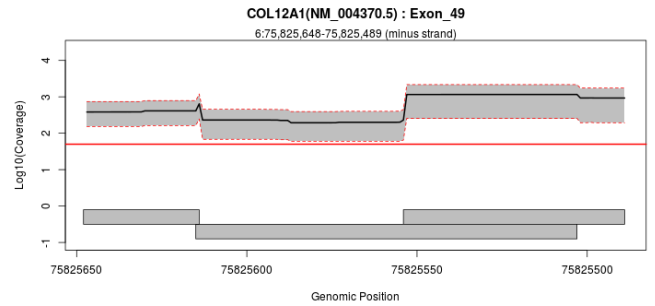
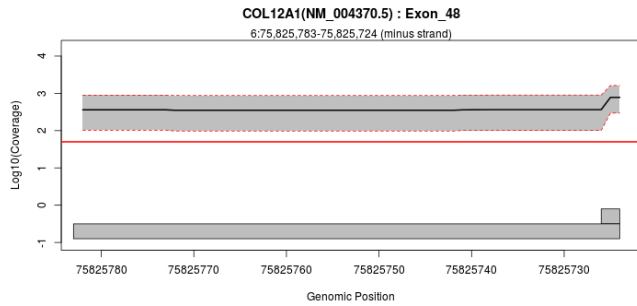
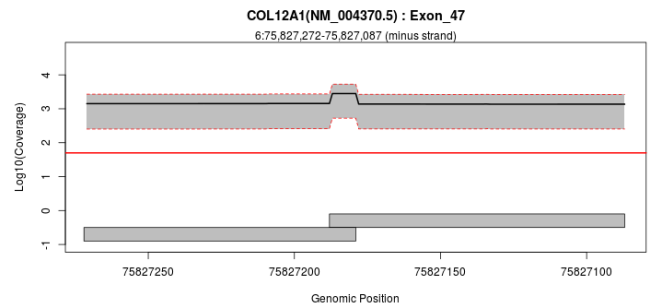
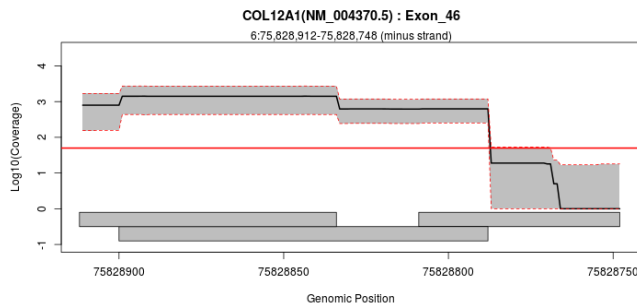
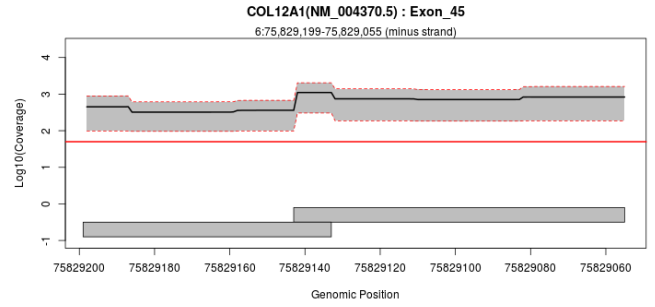
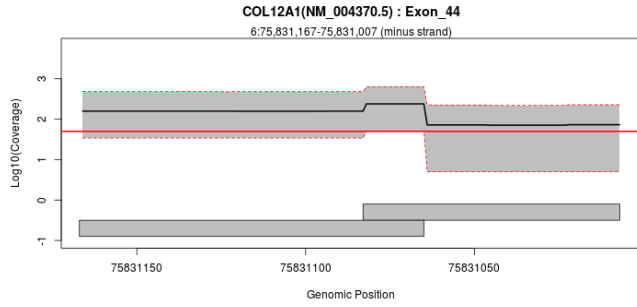
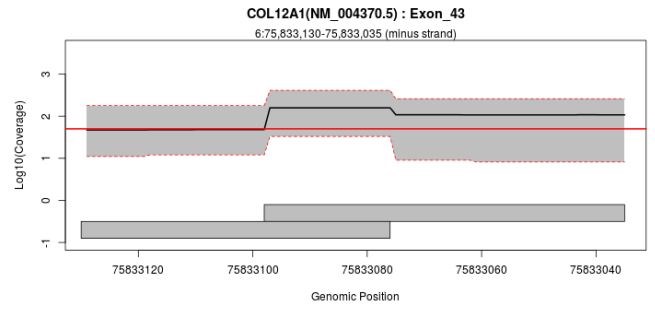
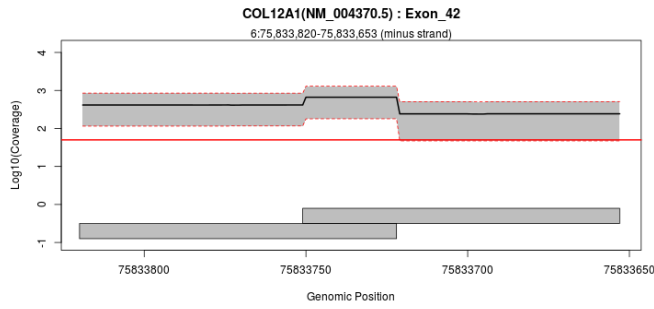
## Exon Overview: COL12A1 : NM\_004370.5

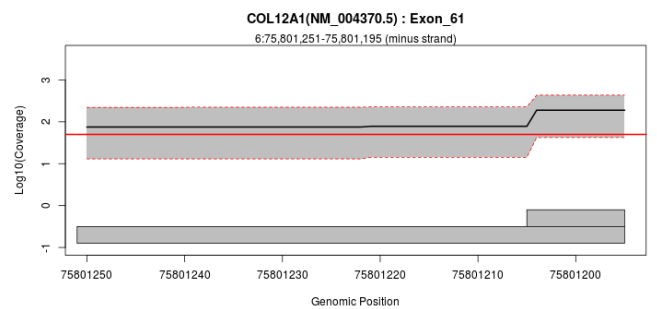
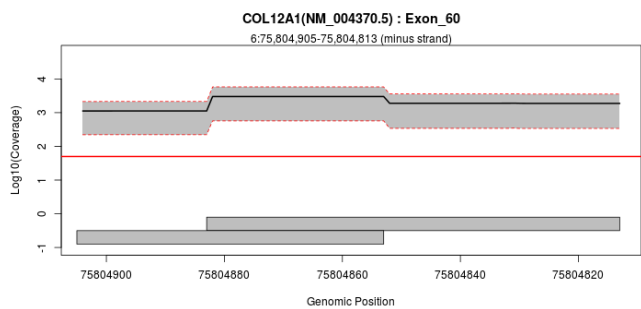
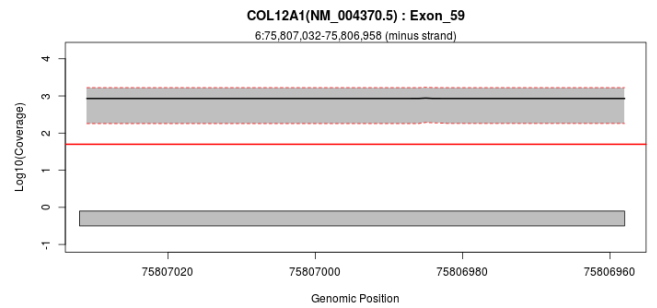
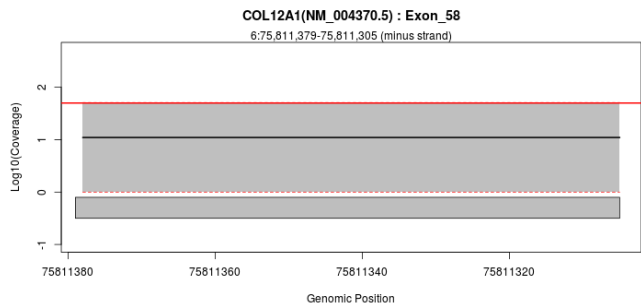
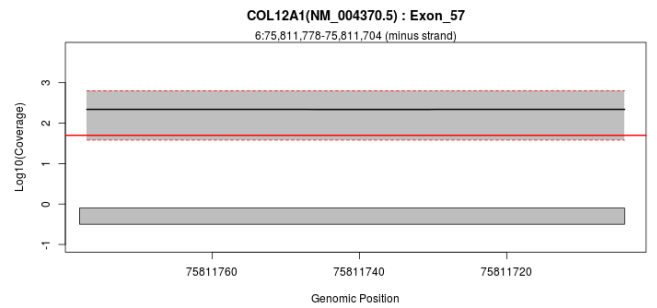
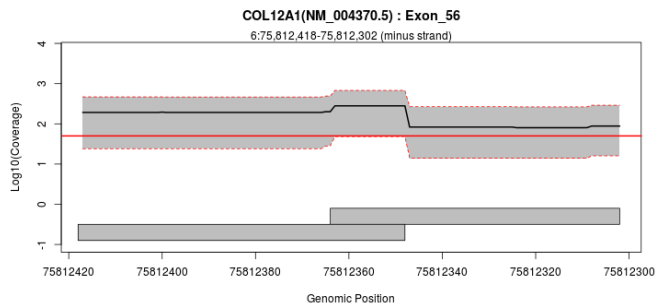
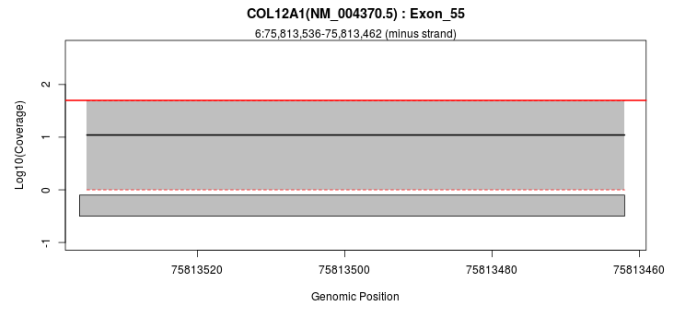
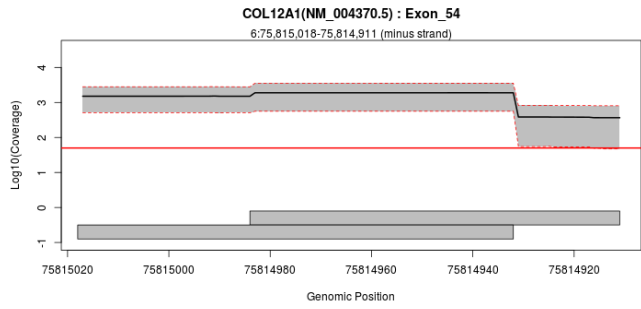
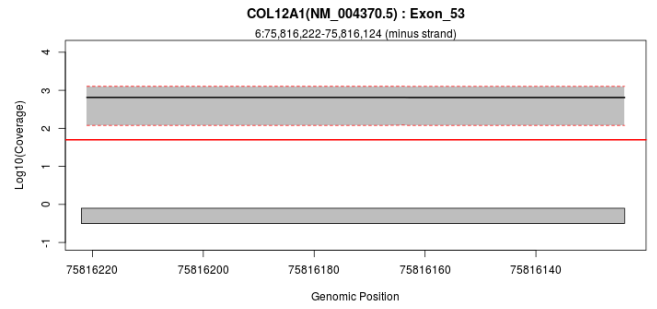
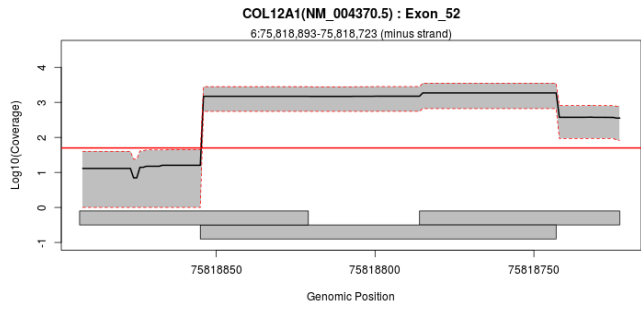


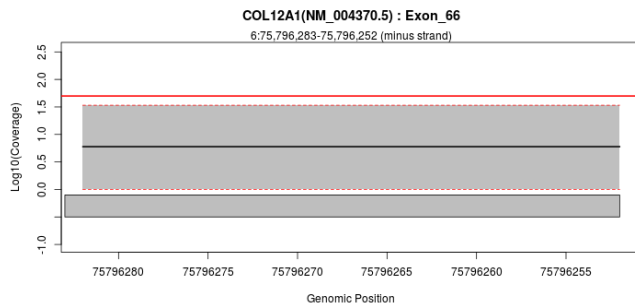
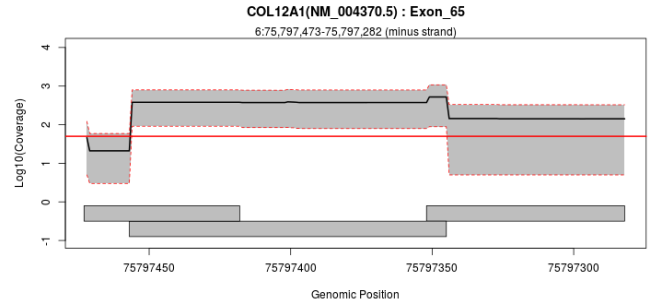
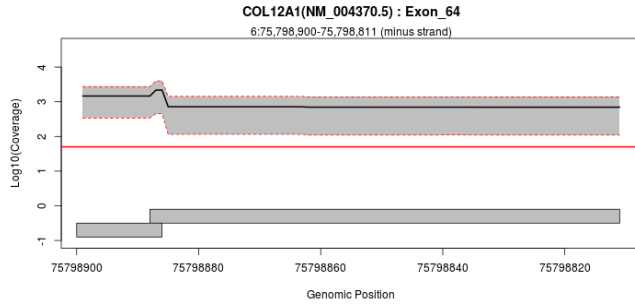
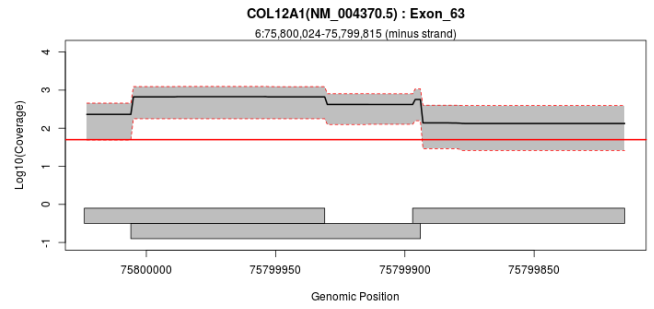
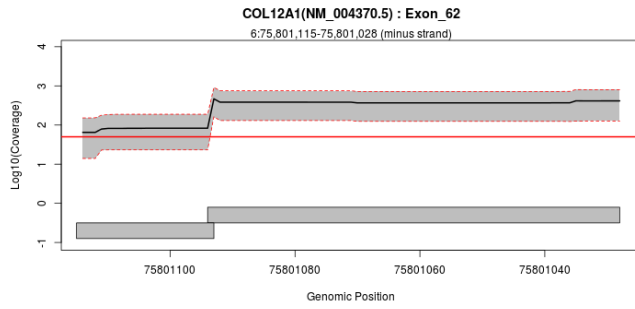




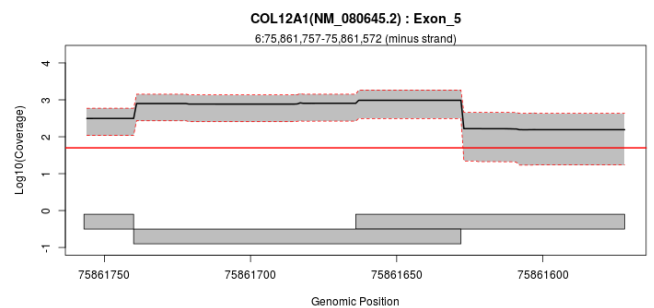
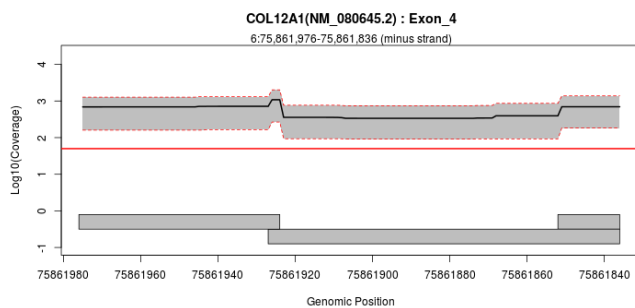
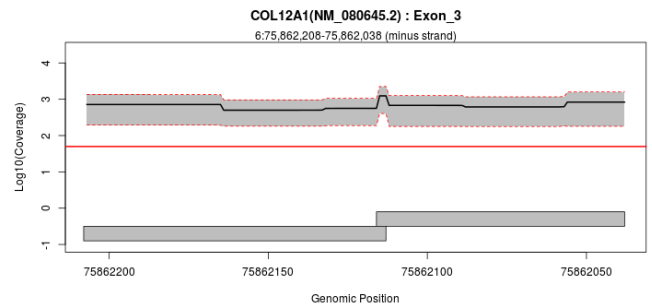
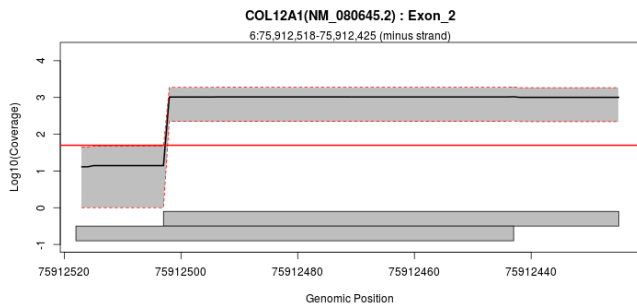


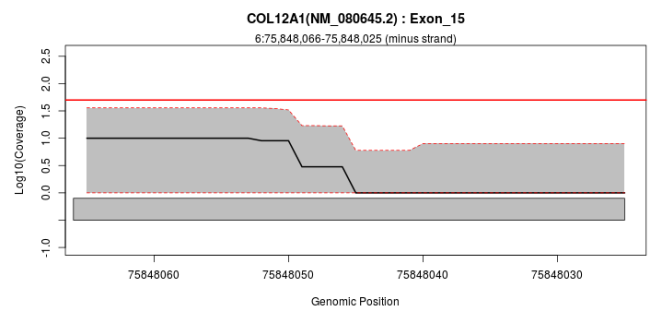
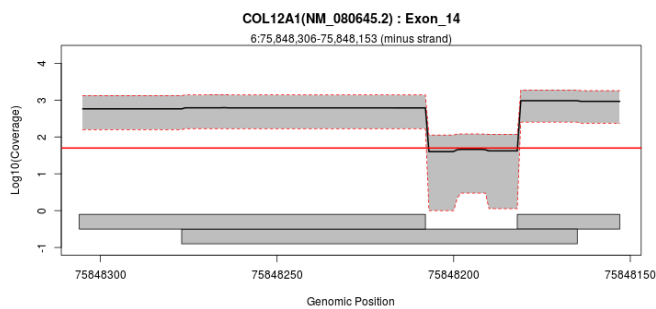
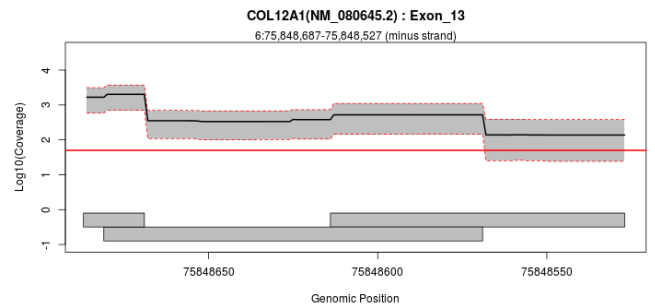
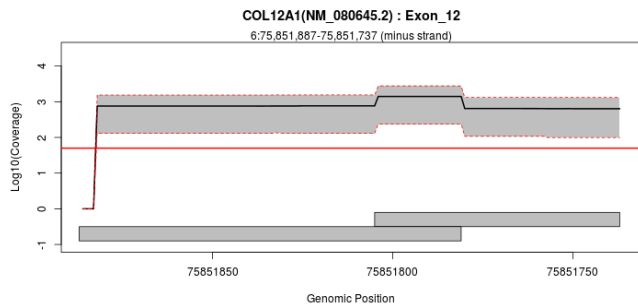
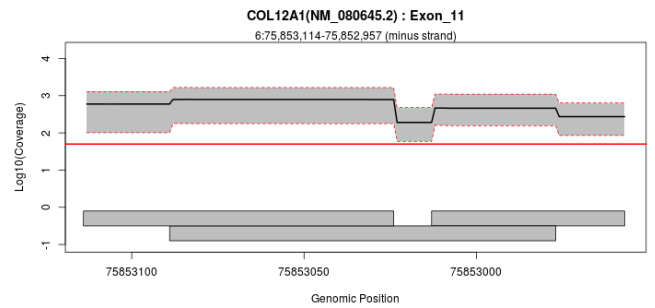
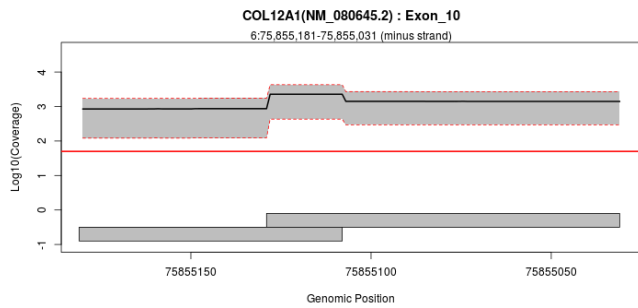
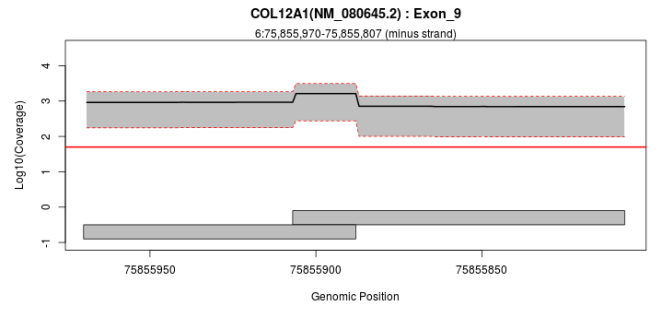
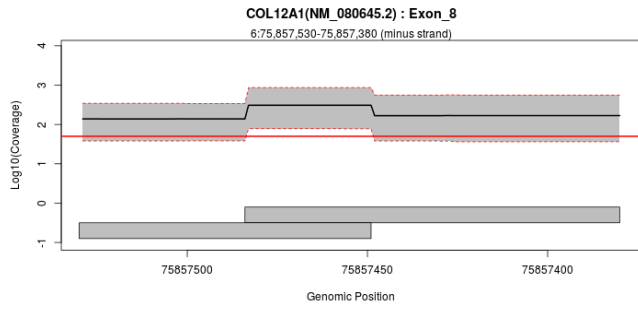
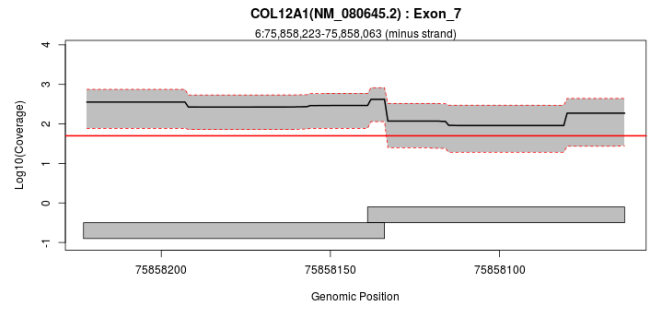
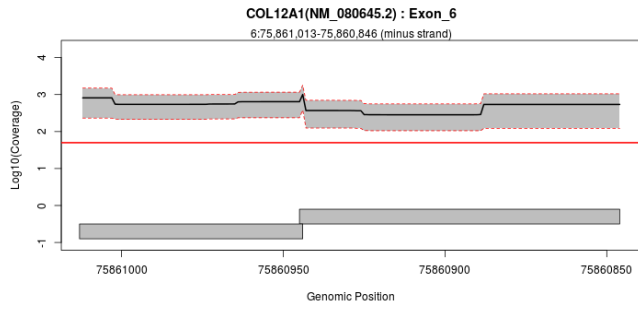




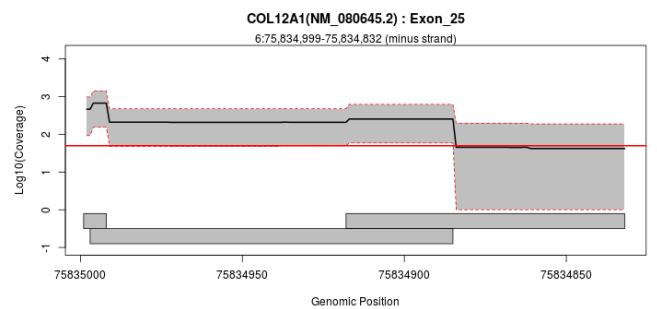
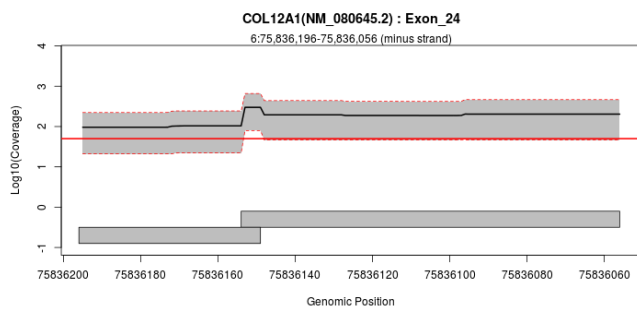
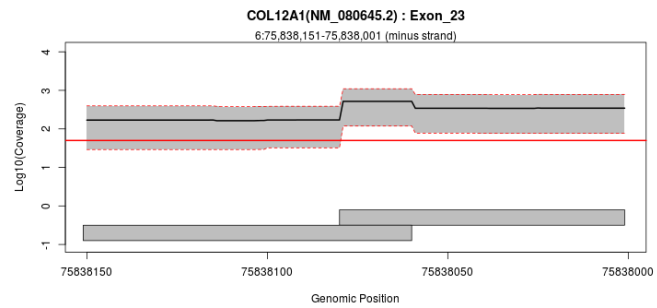
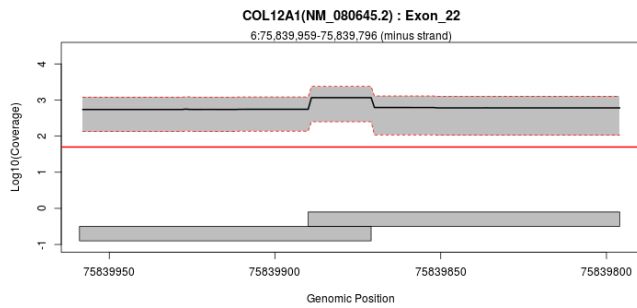
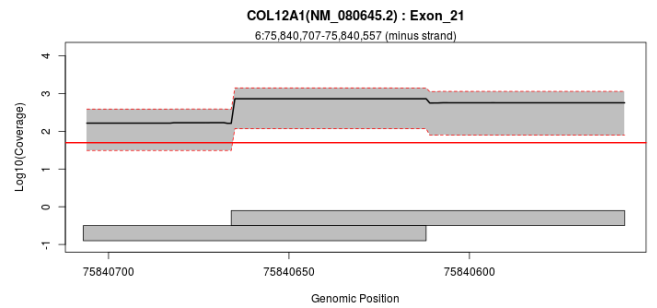
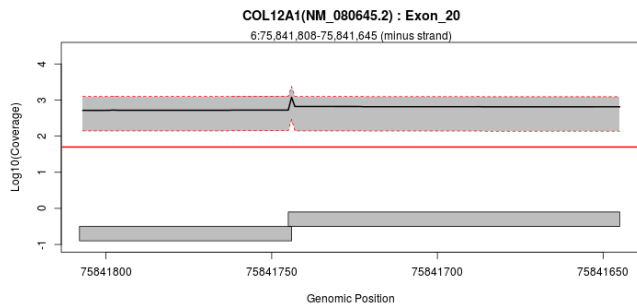
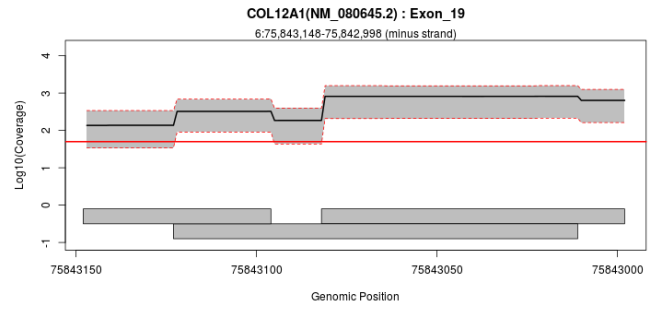
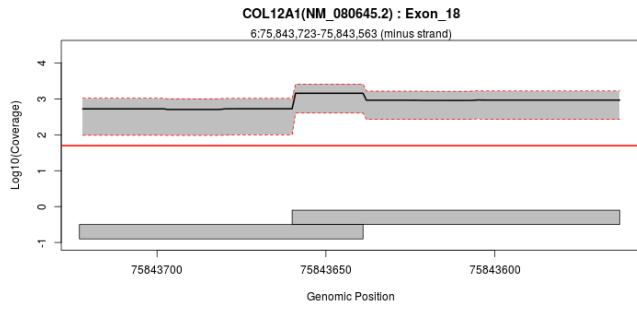
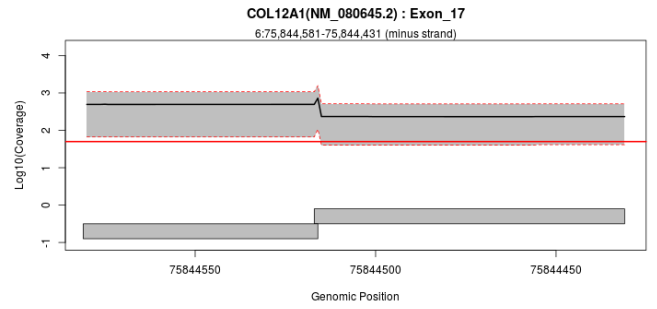
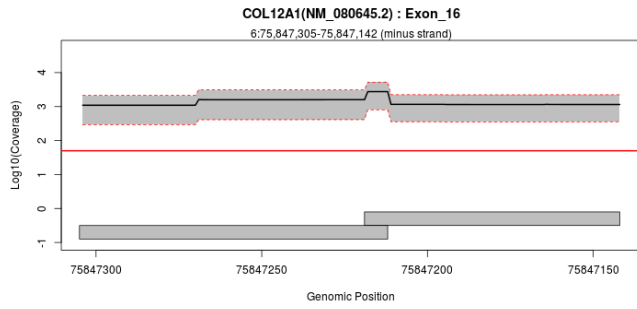


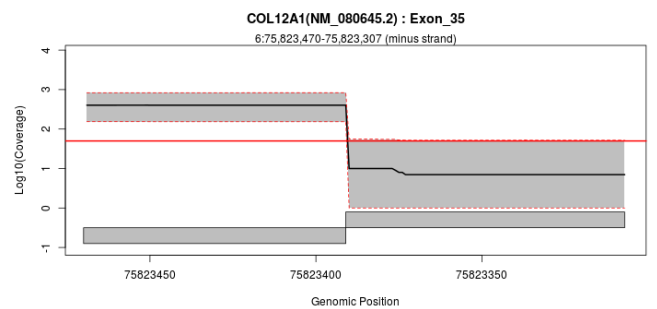
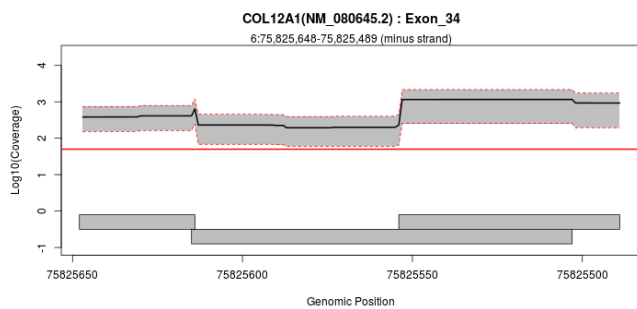
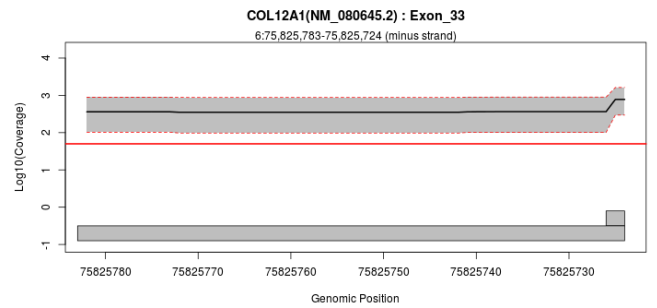
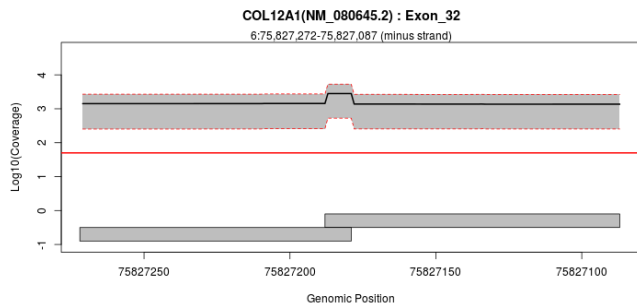
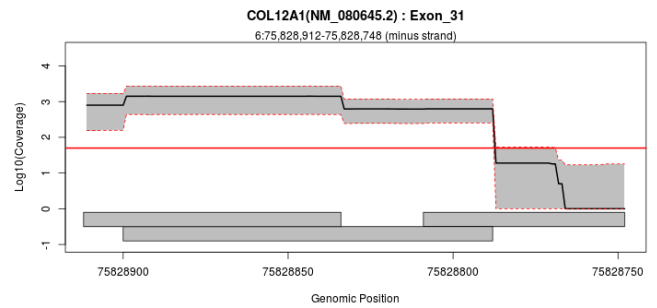
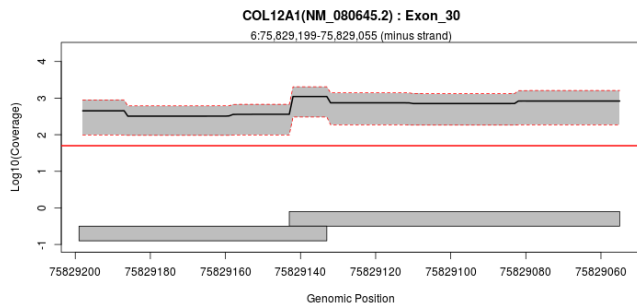
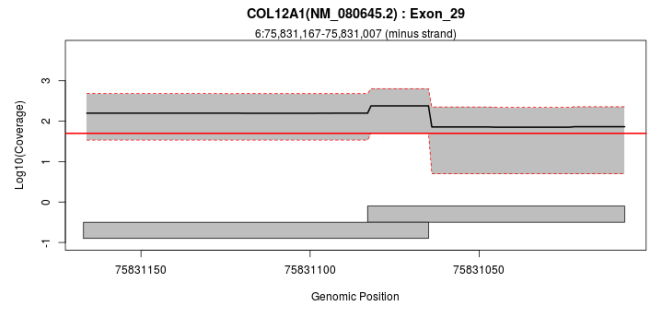
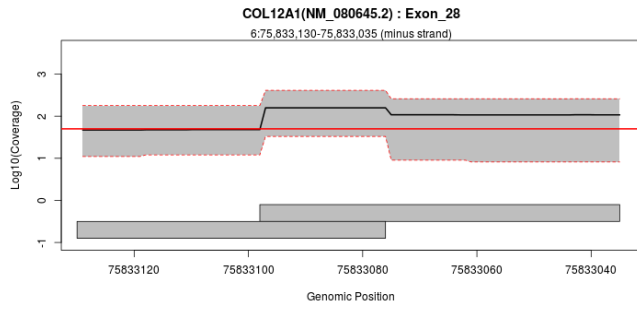
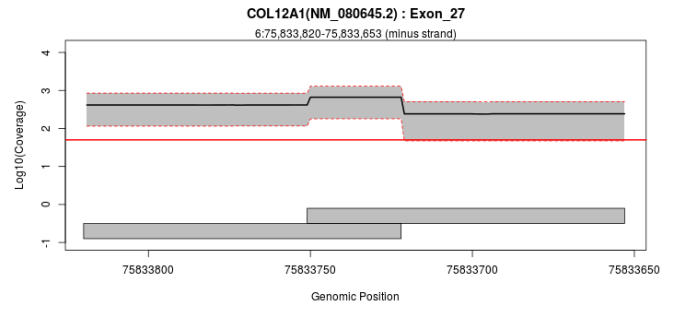
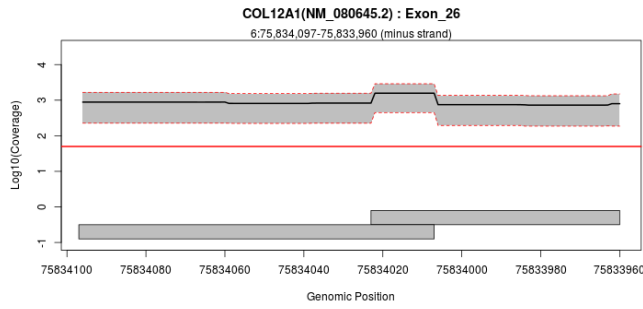
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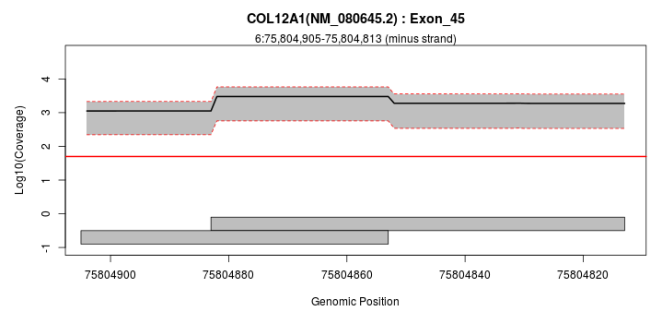
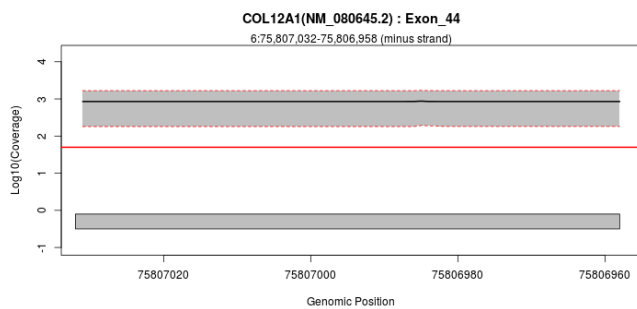
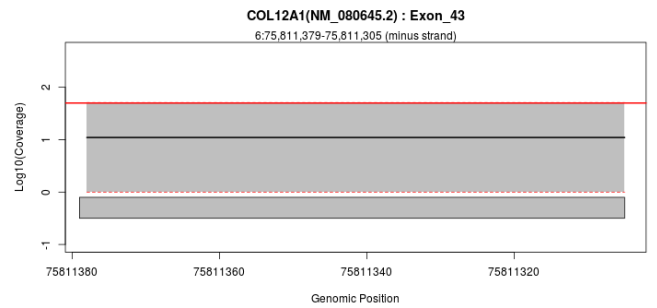
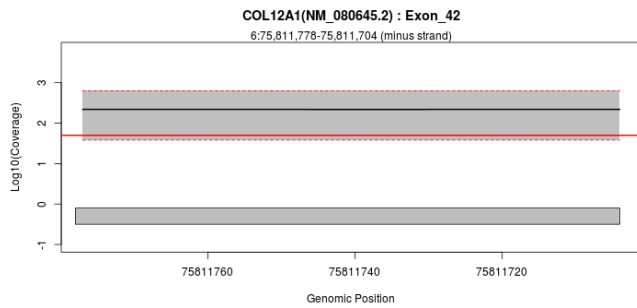
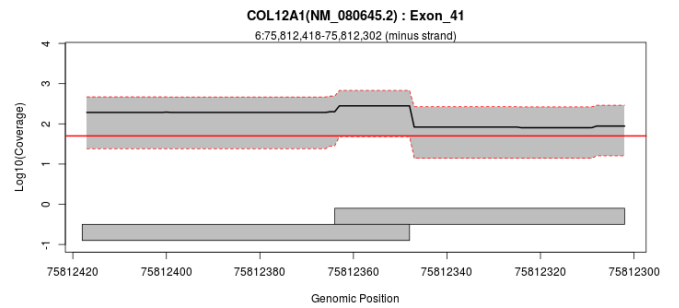
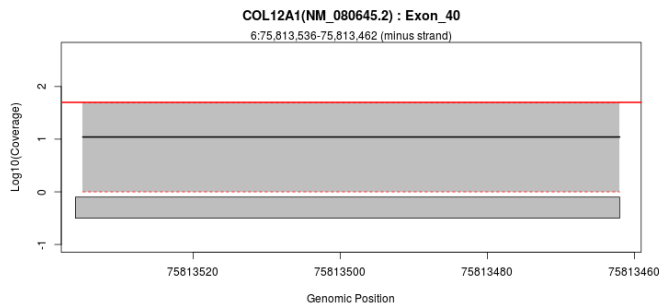
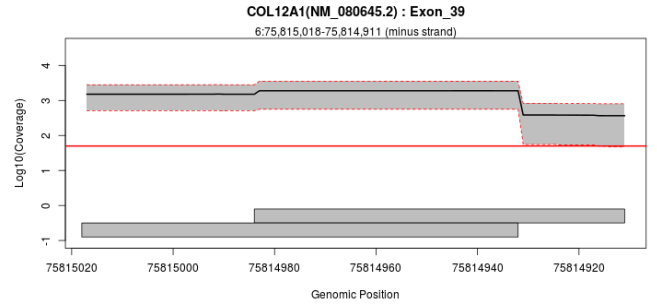
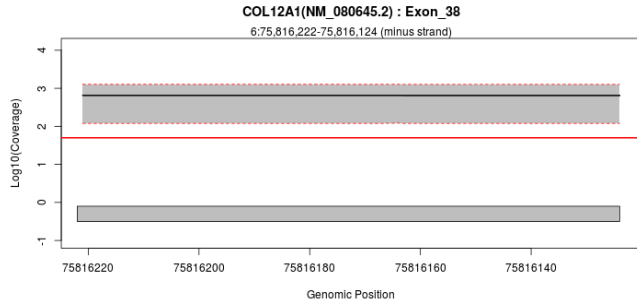
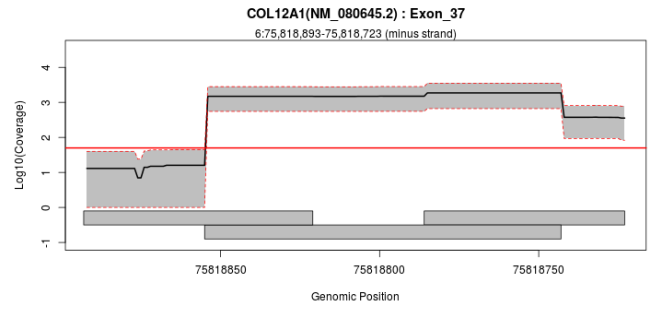
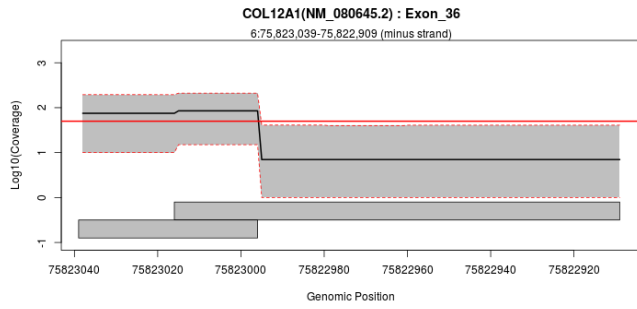


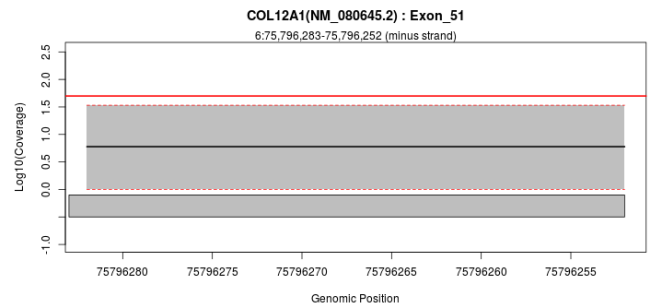
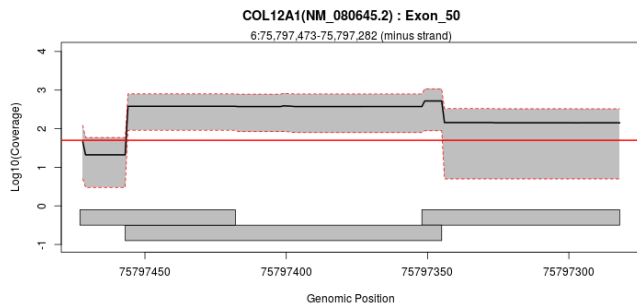
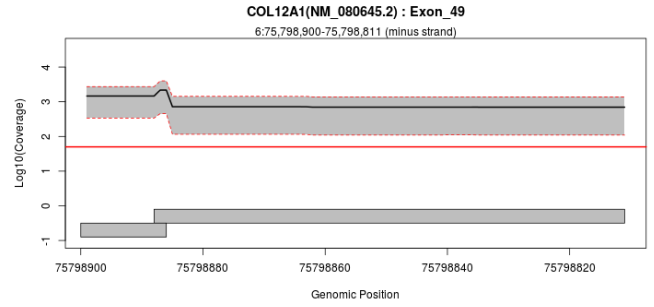
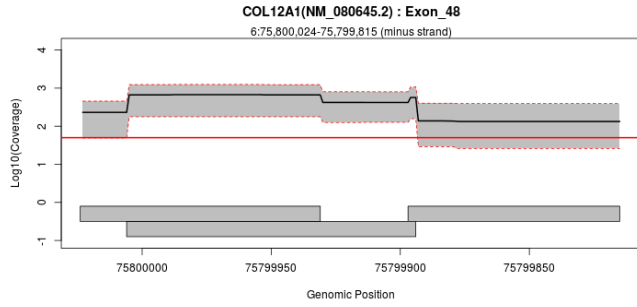
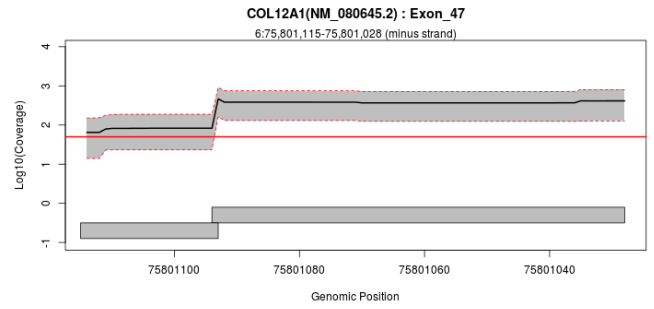
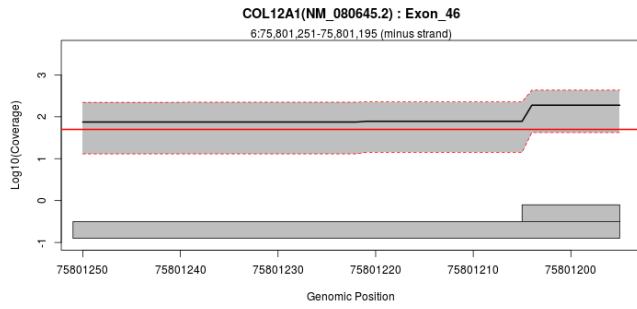




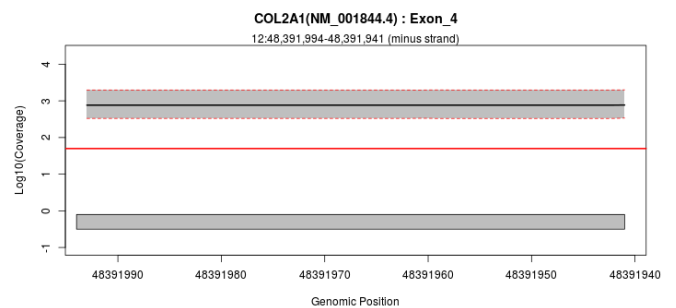
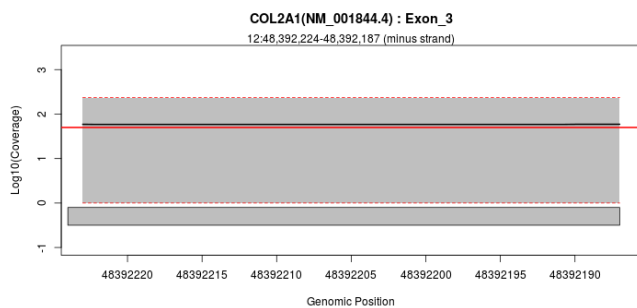
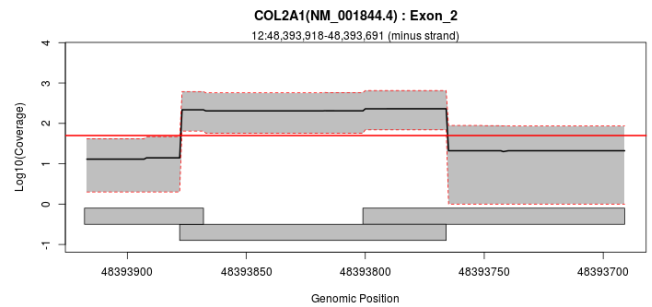
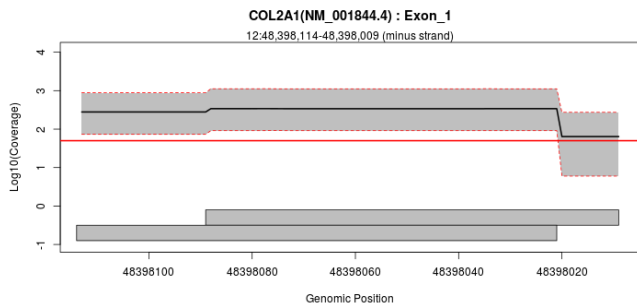


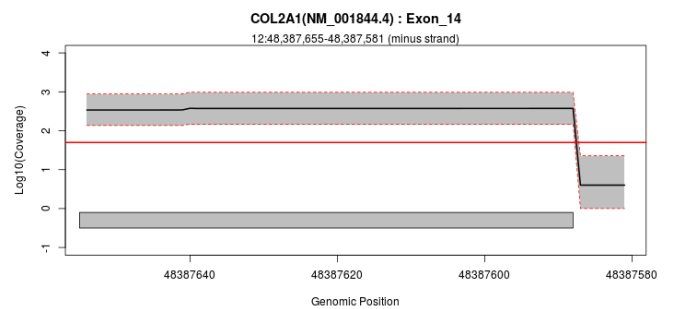
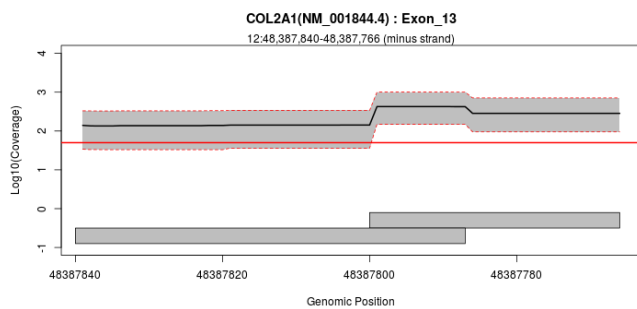
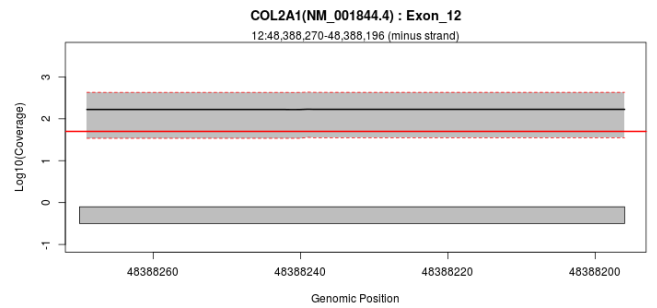
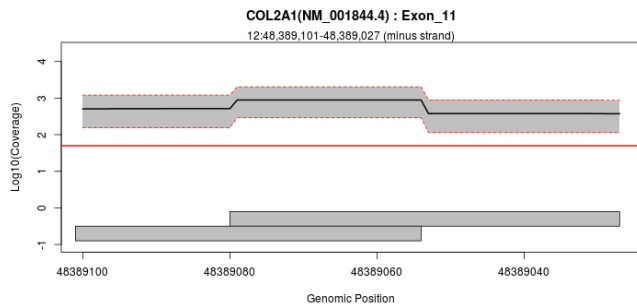
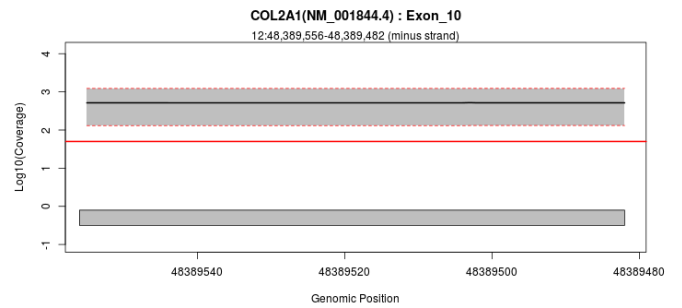
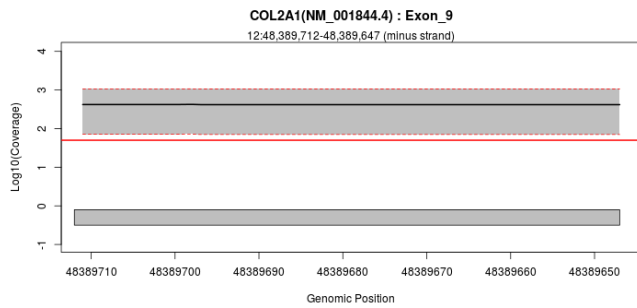
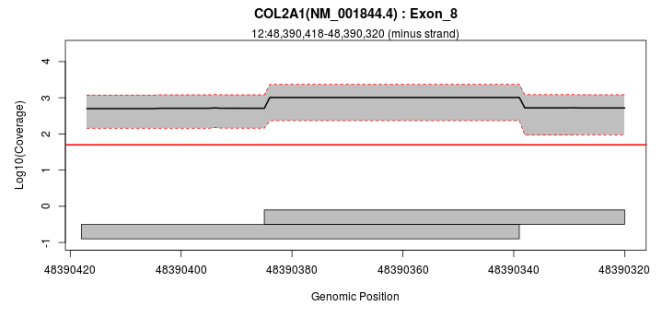
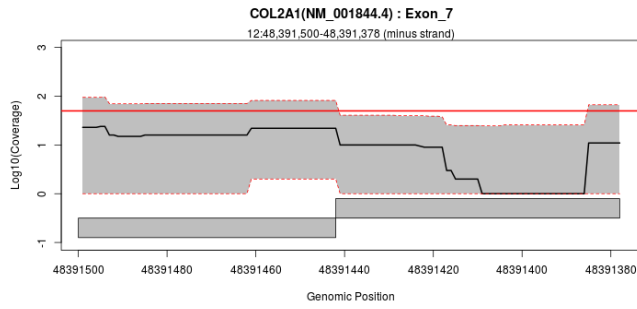
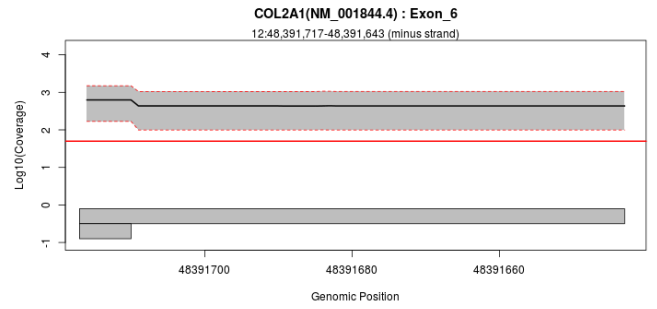
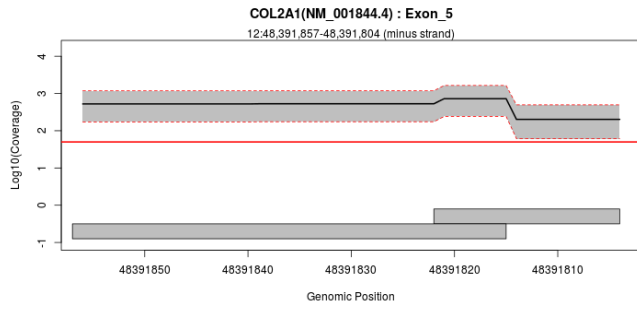


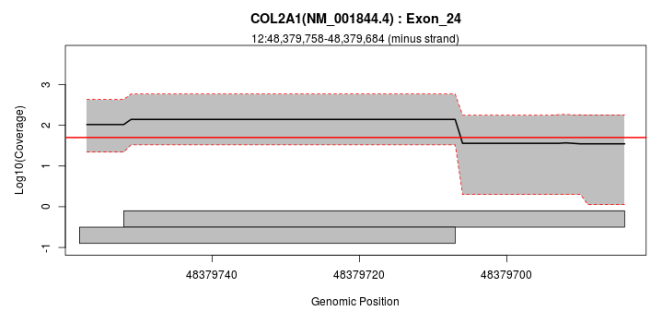
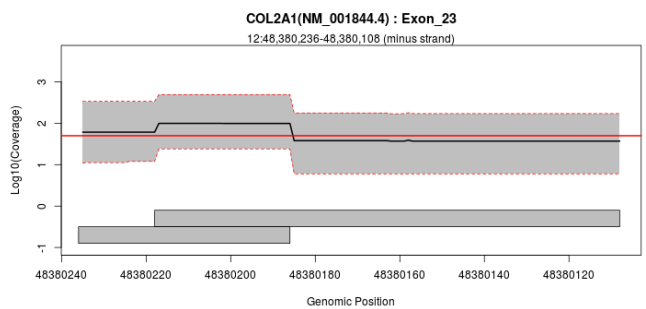
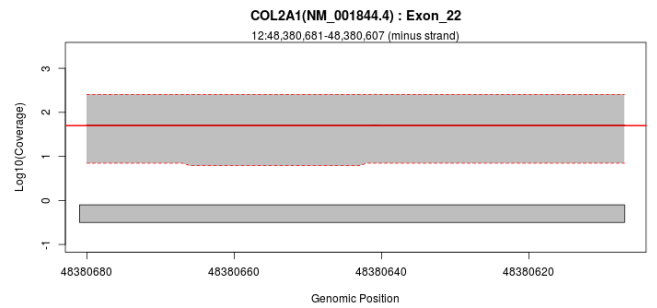
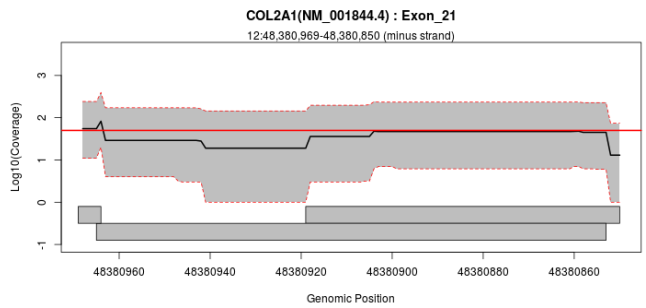
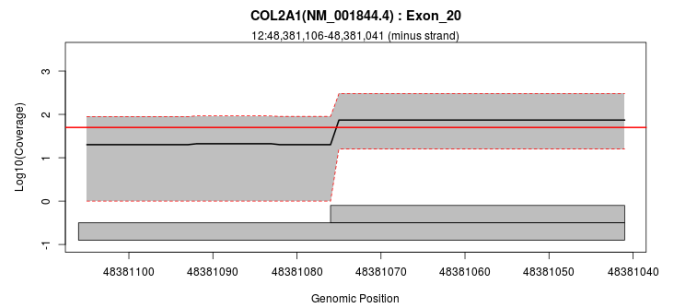
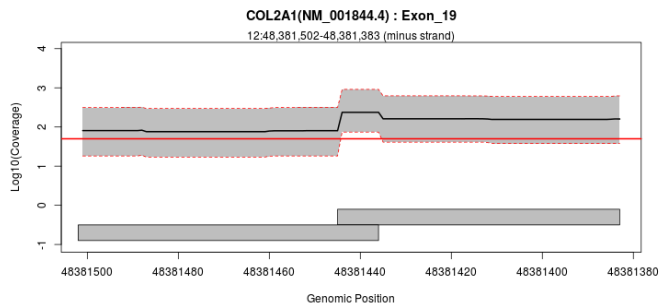
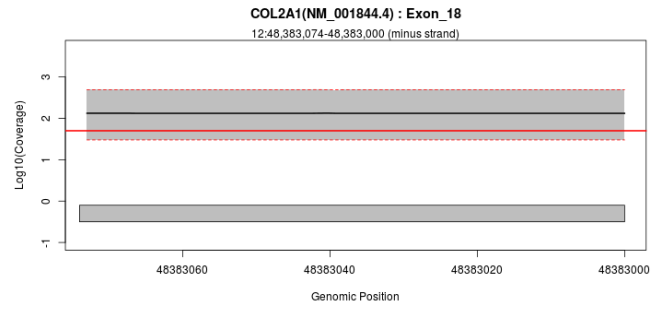
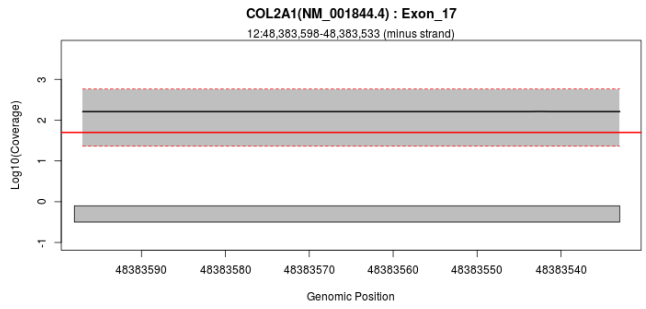
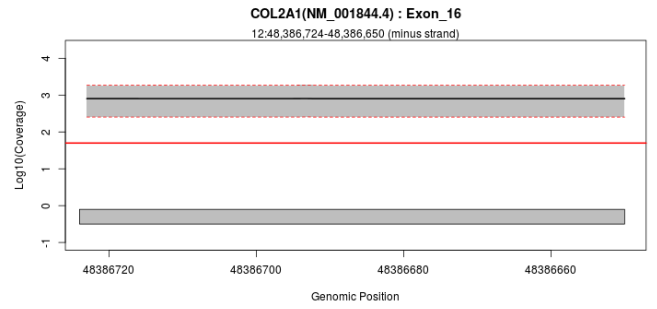
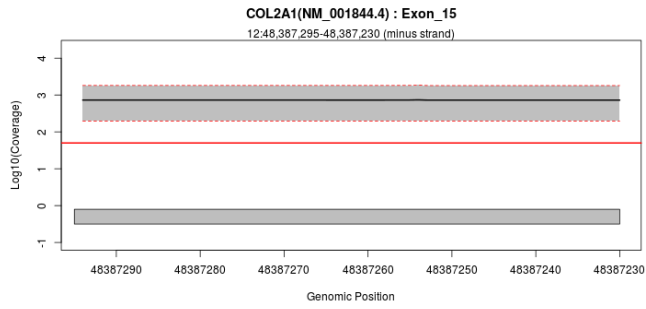


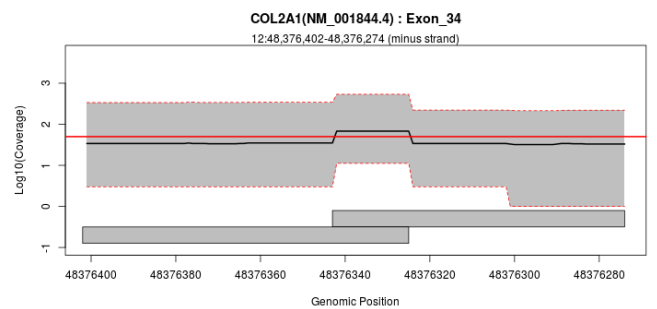
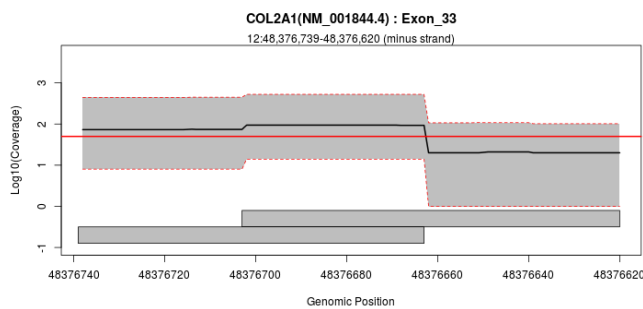
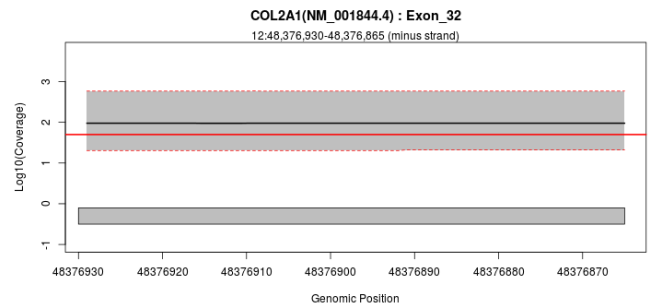
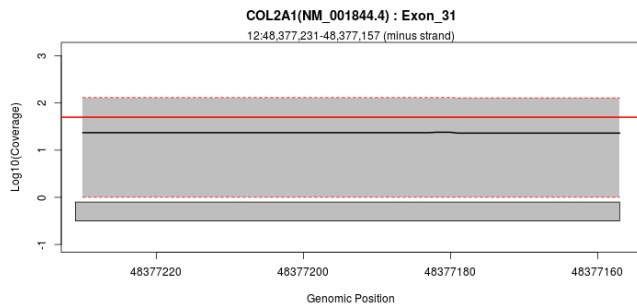
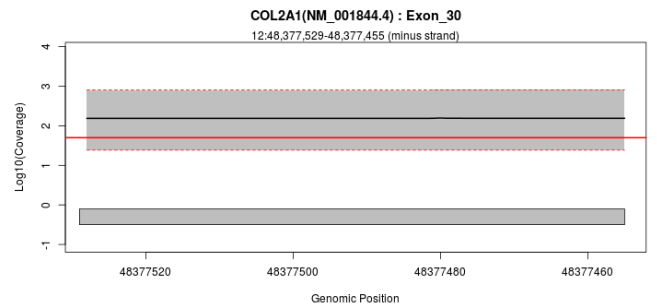
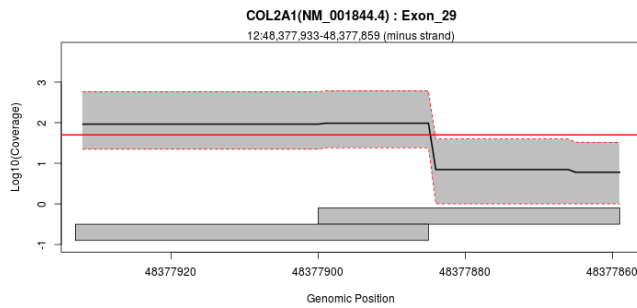
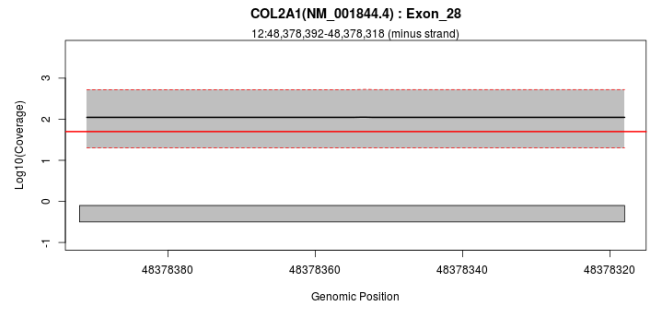
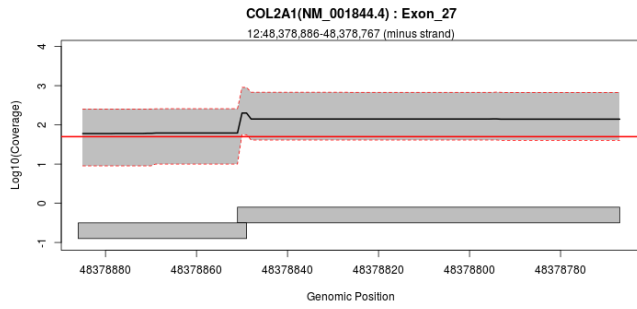
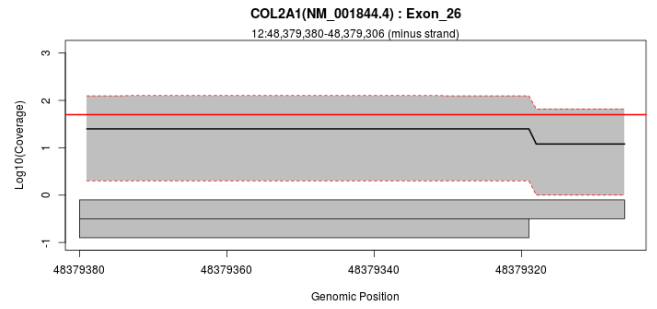
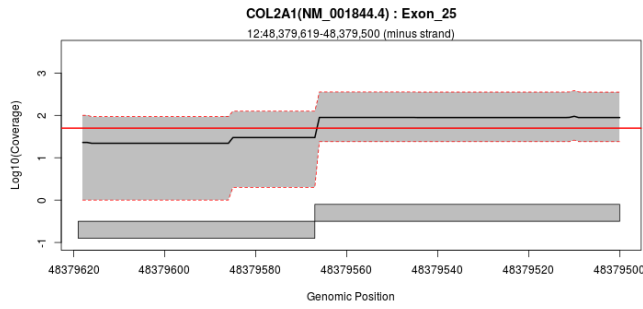


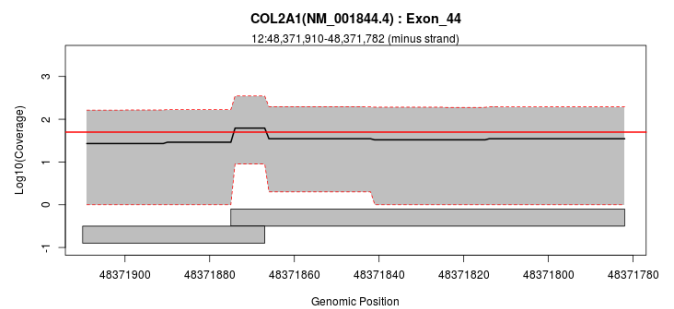
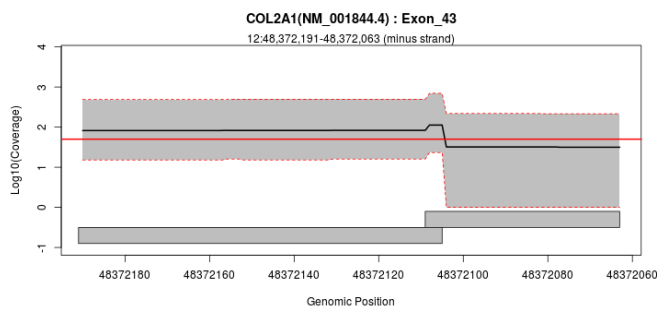
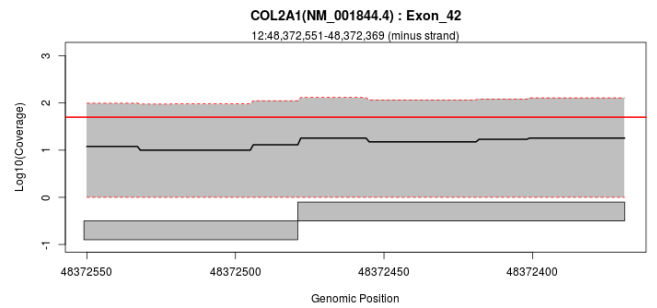
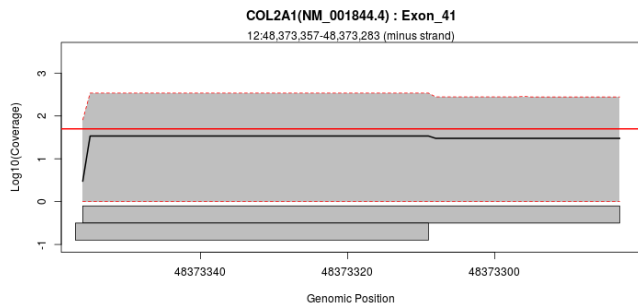
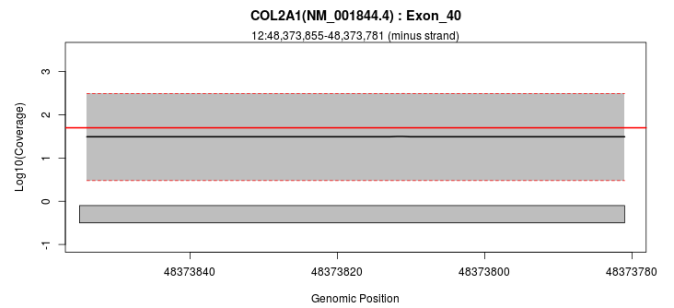
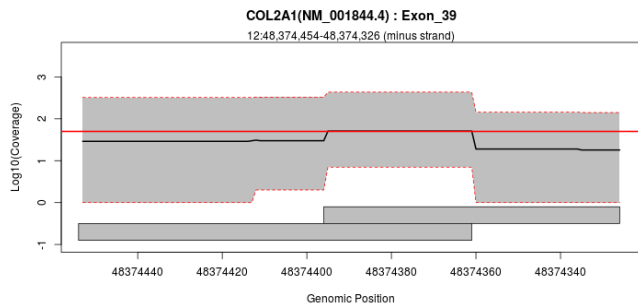
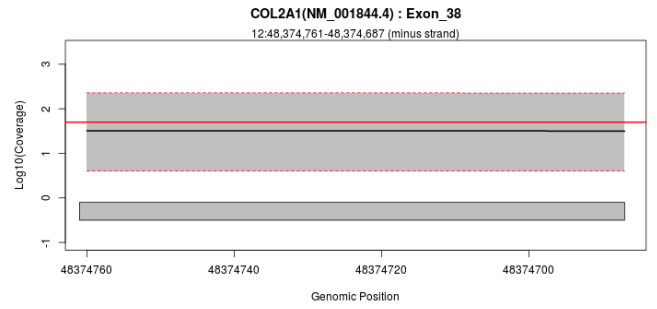
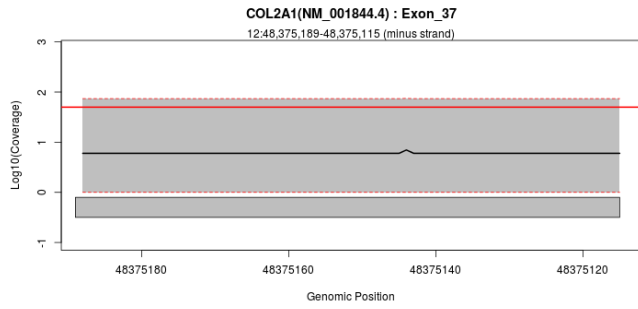
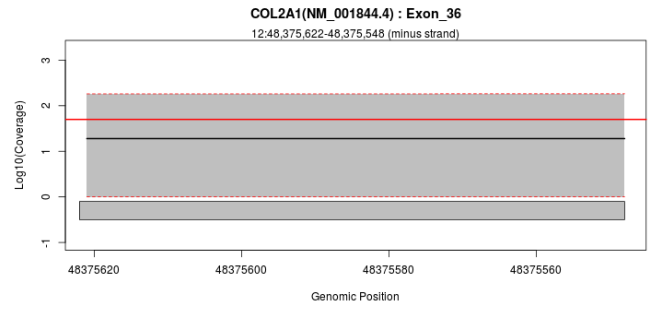
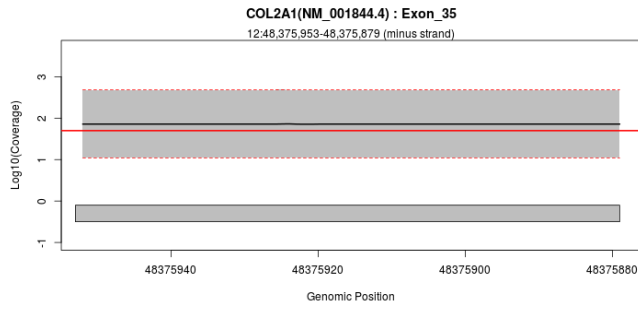
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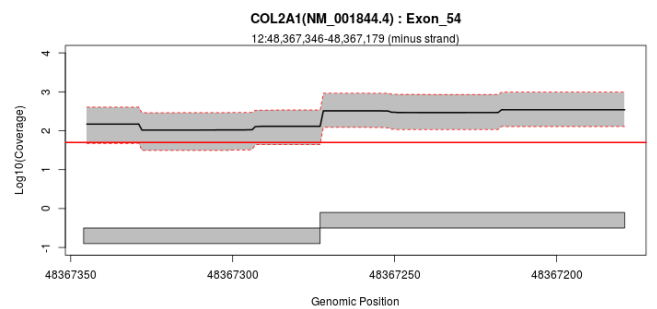
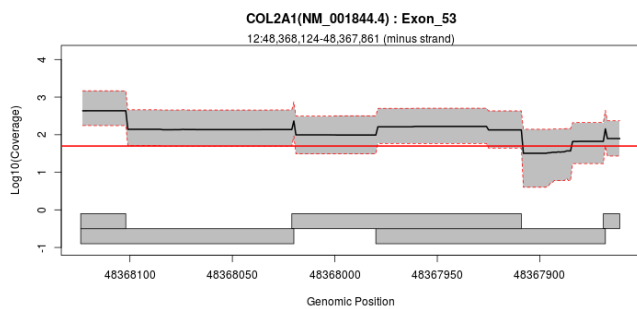
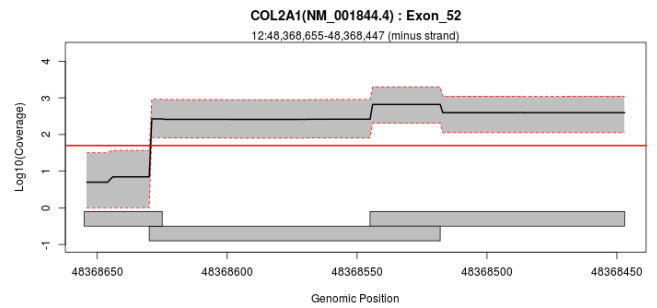
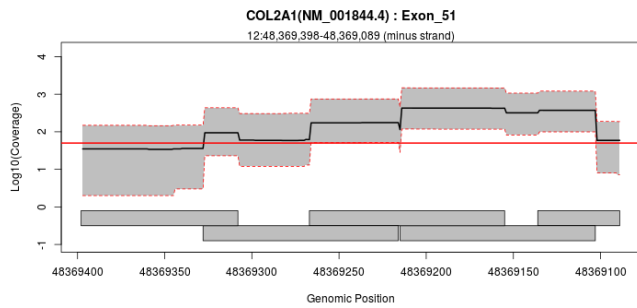
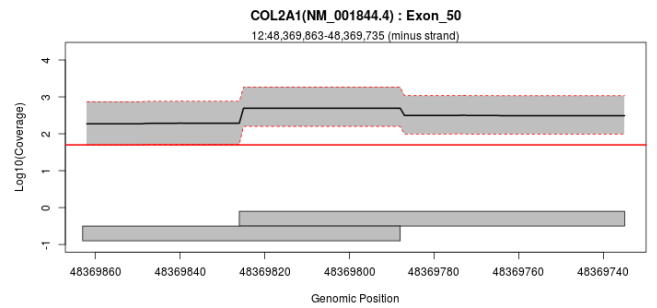
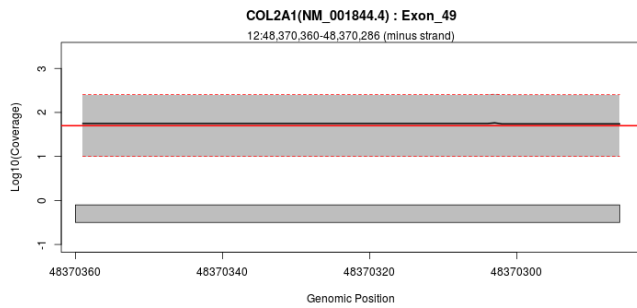
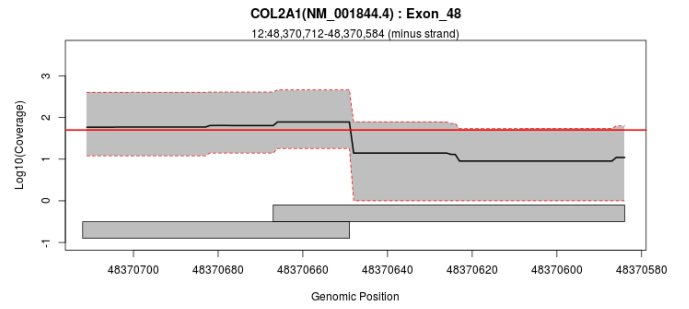
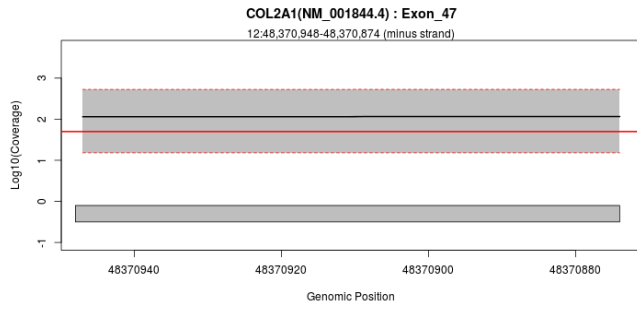
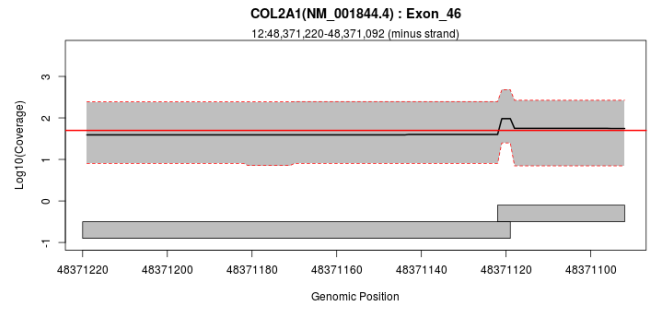
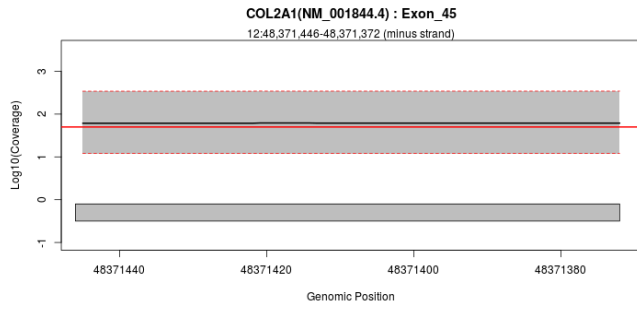




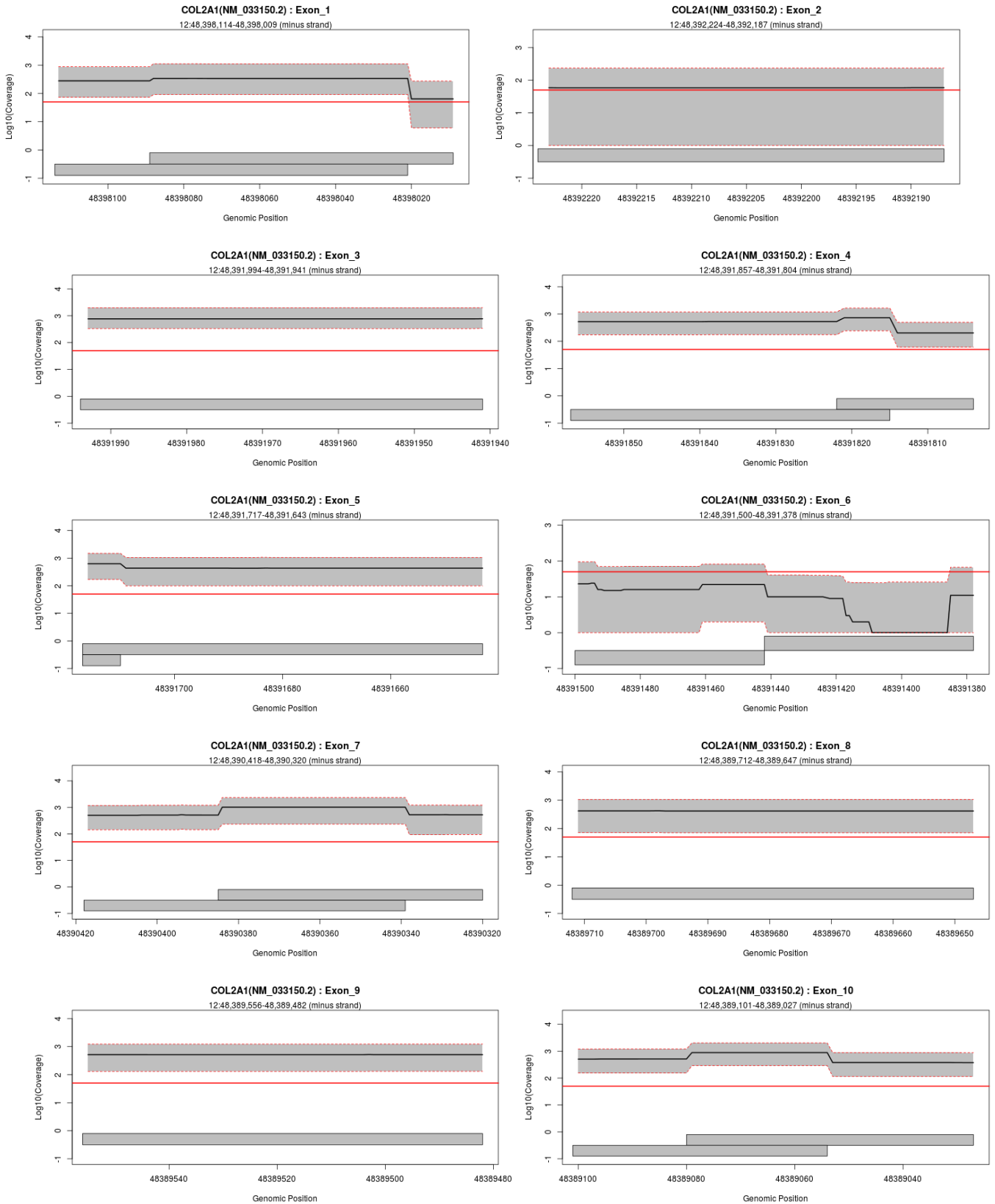


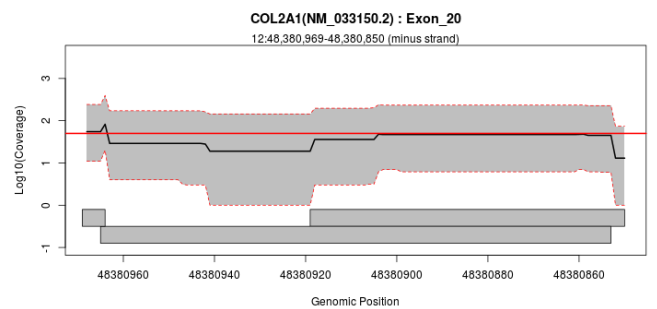
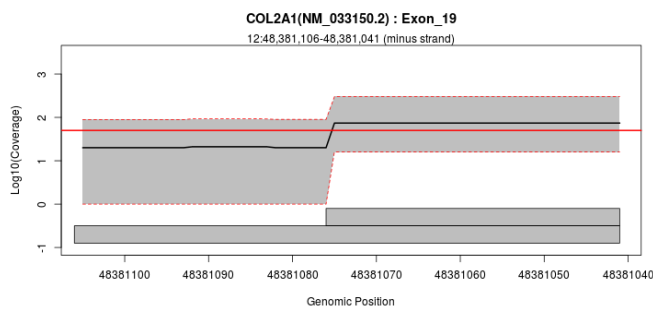
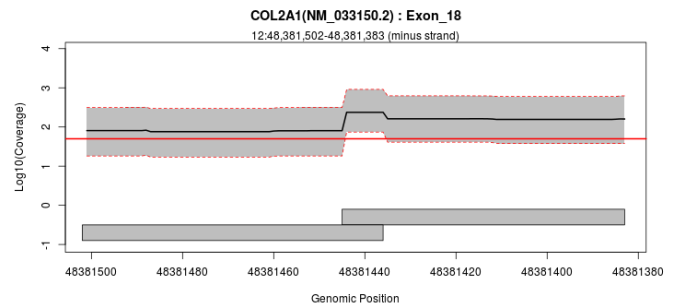
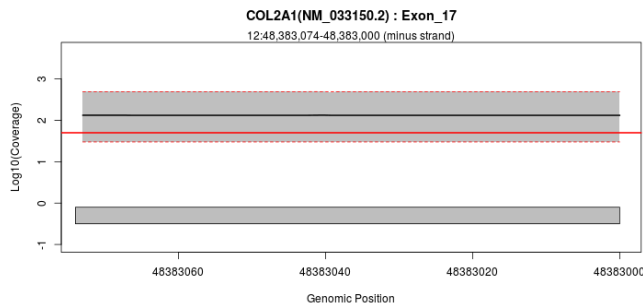
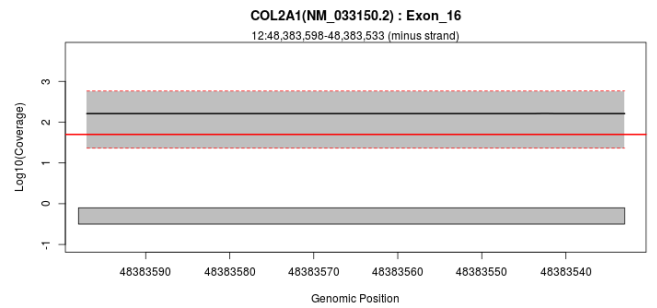
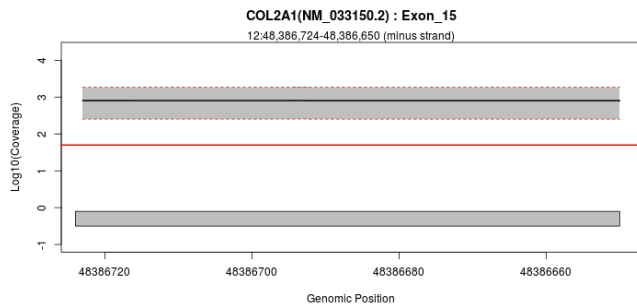
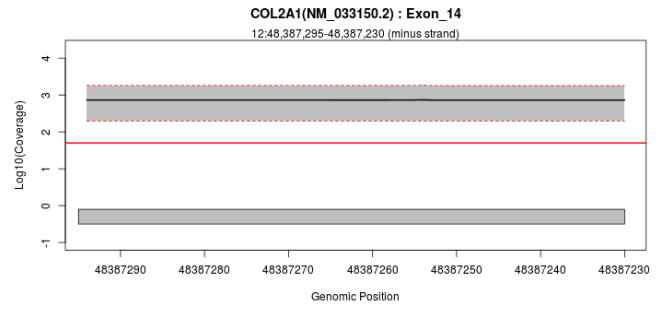
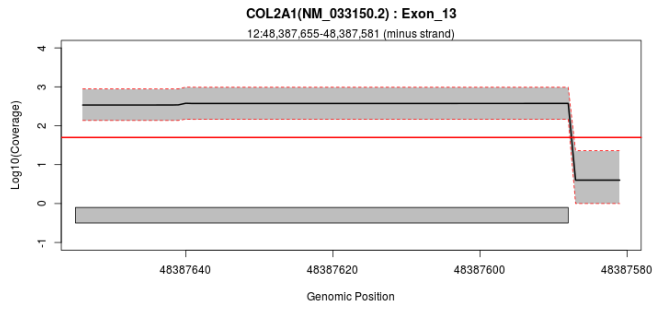
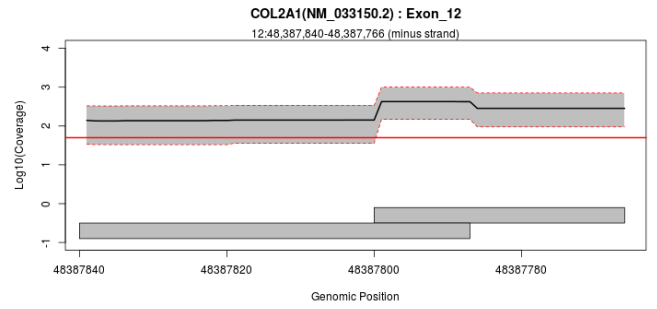
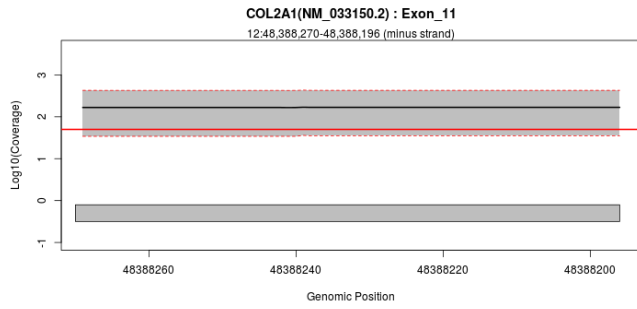


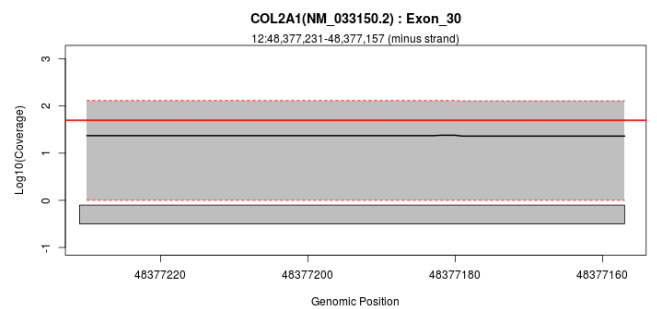
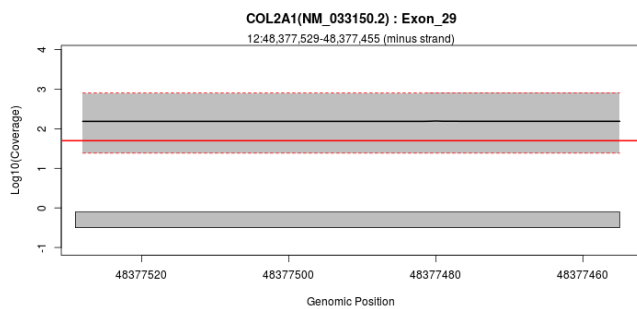
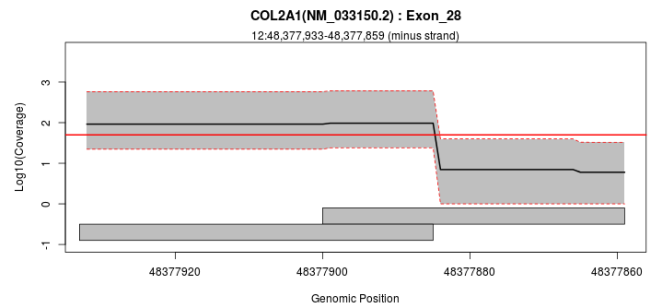
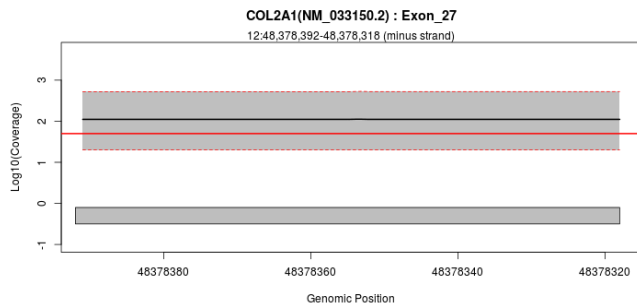
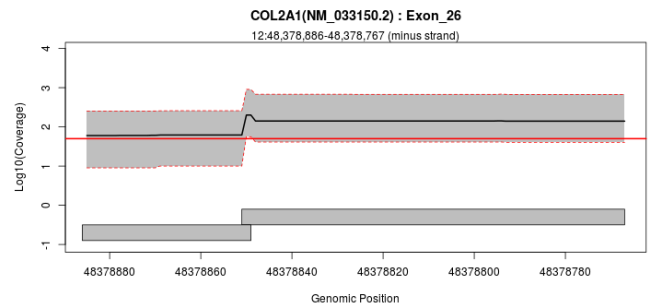
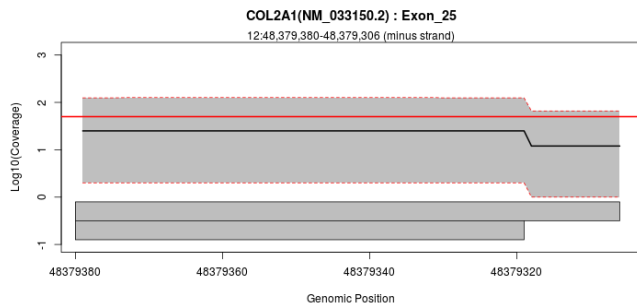
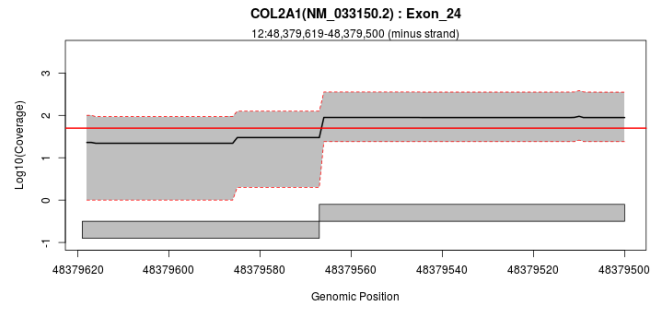
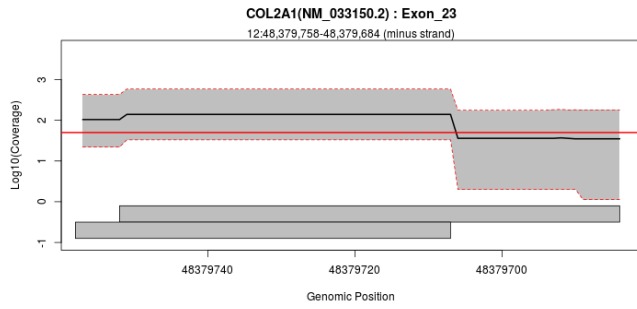
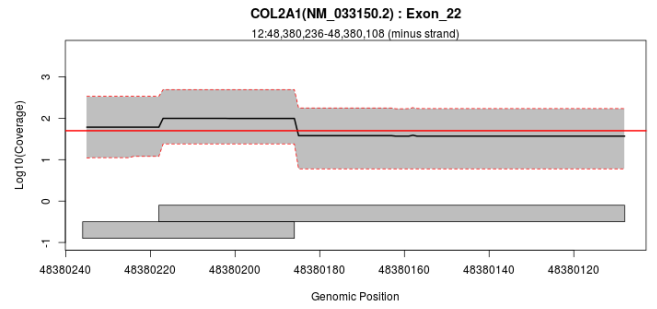
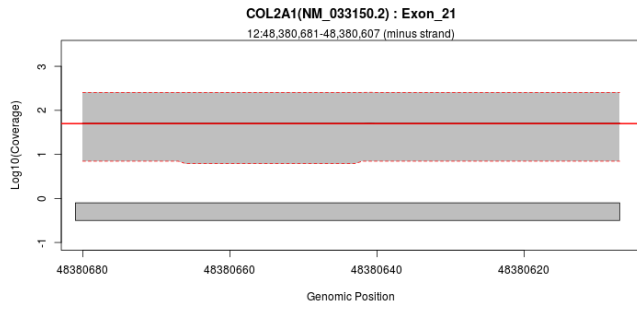


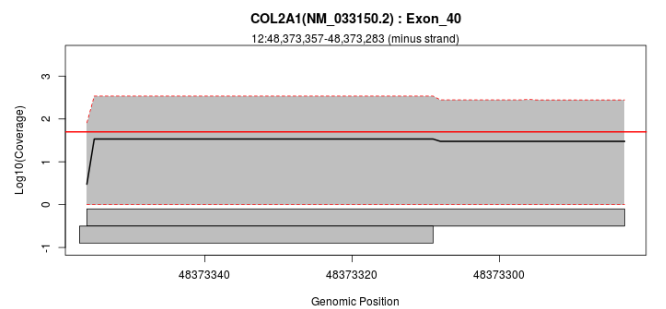
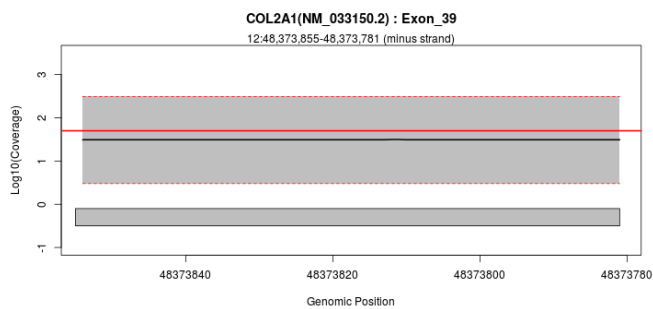
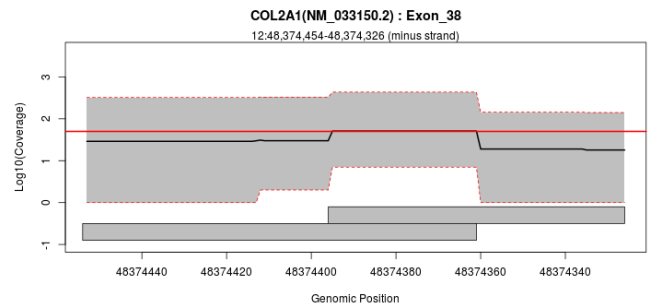
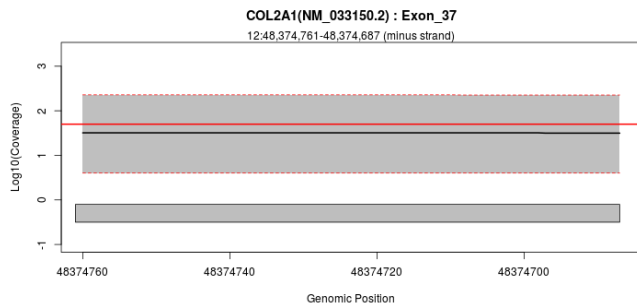
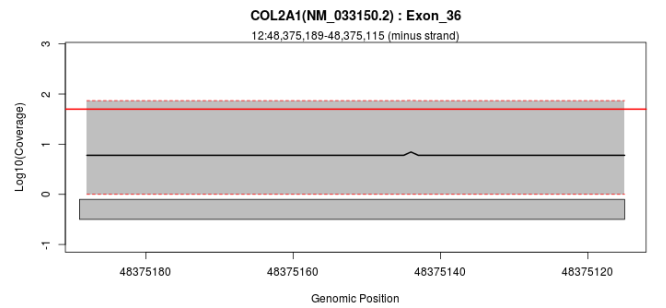
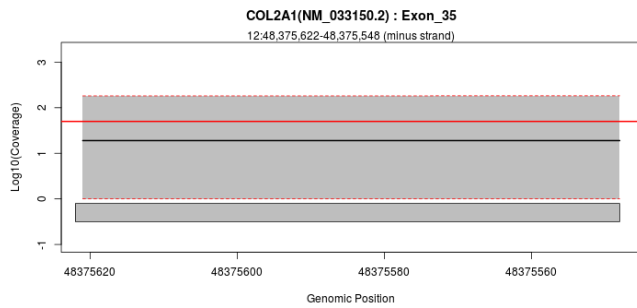
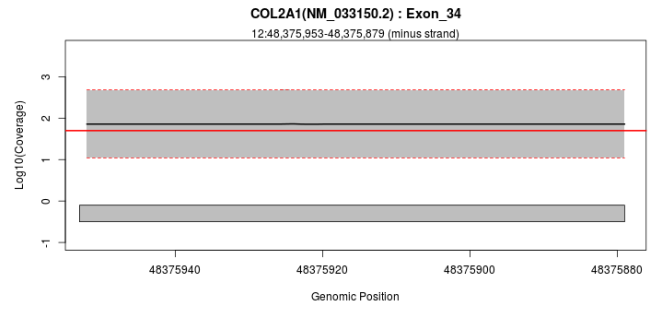
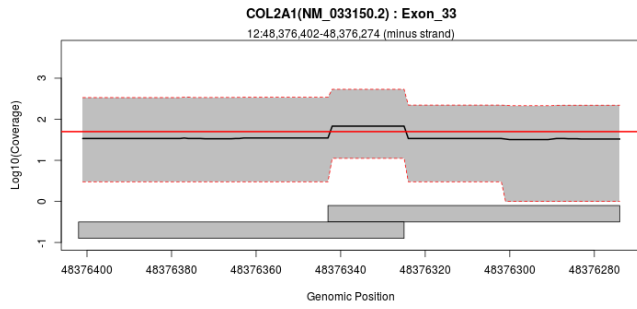
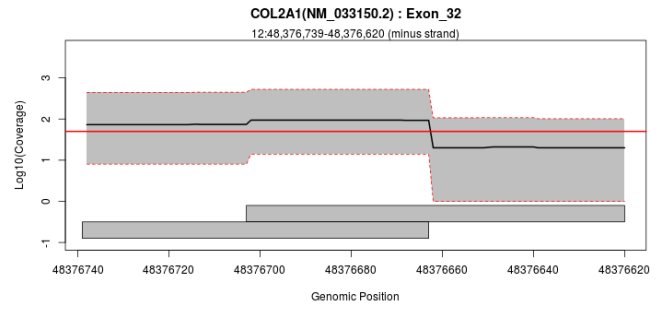
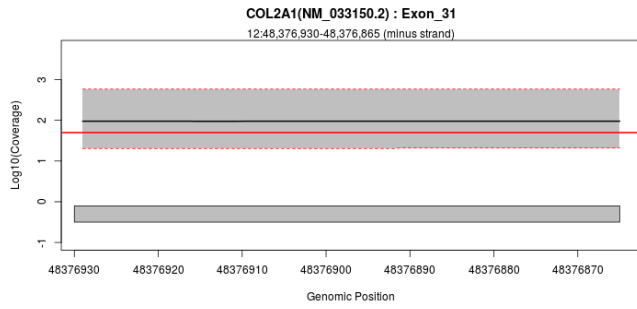


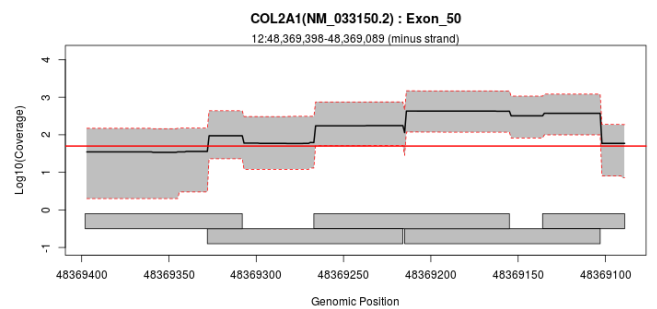
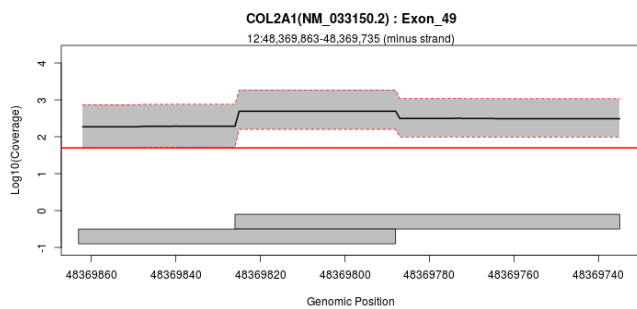
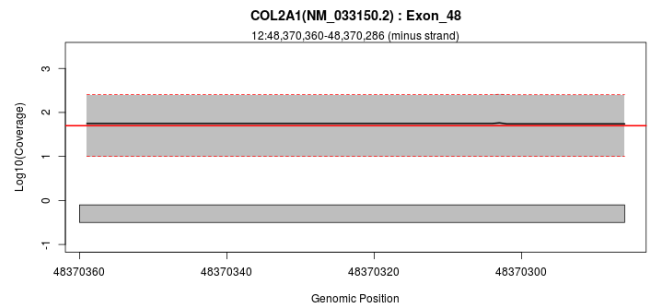
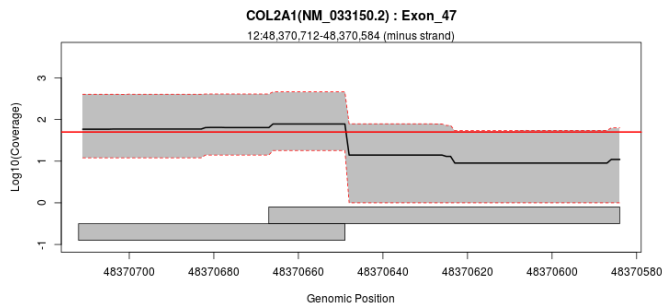
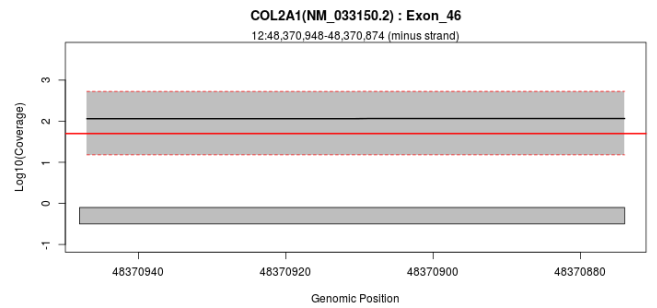
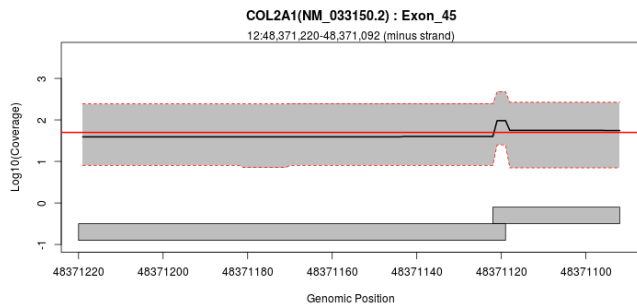
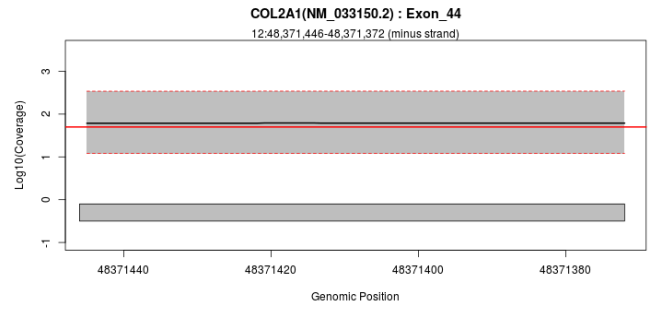
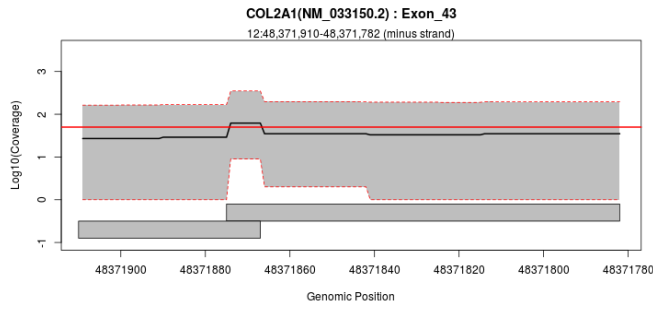
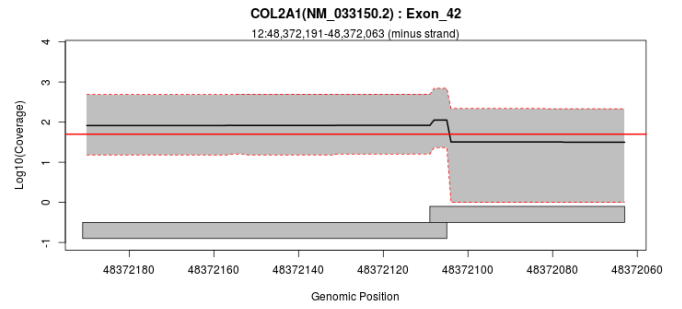
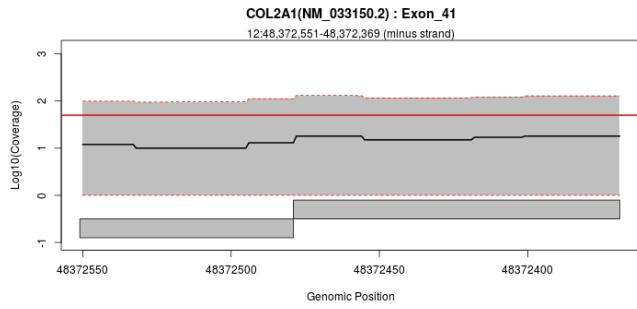
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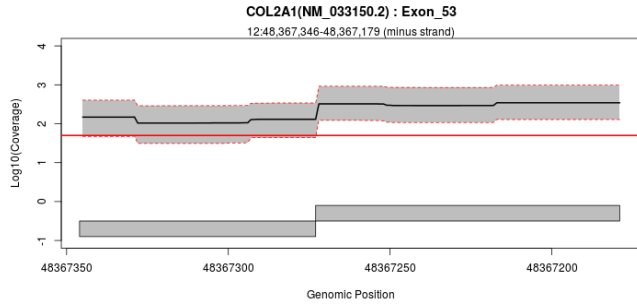
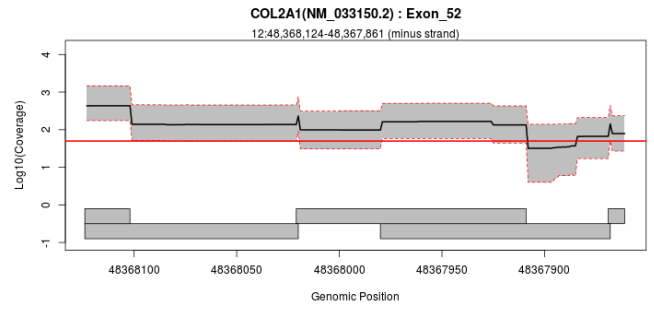
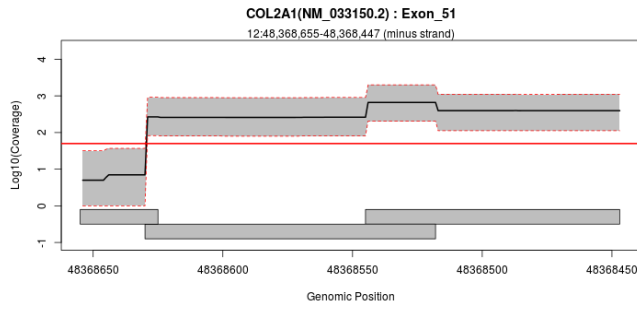




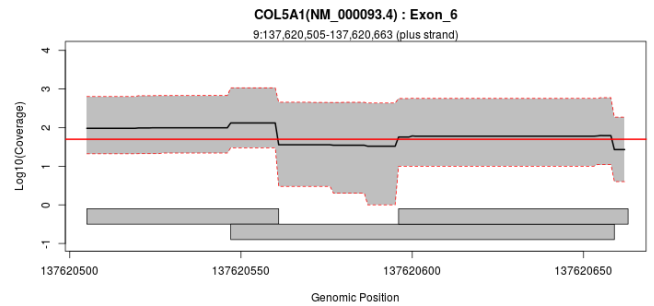
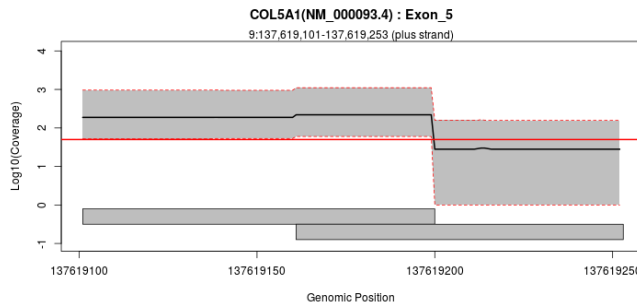
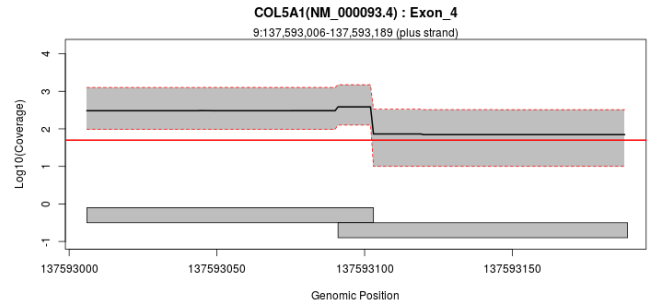
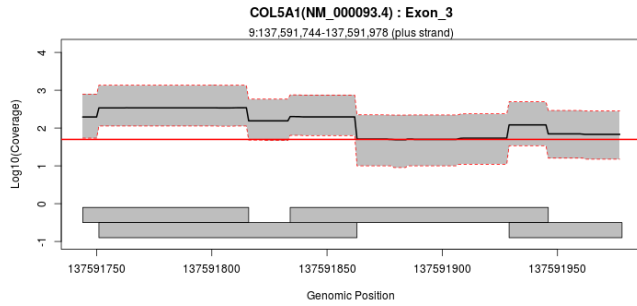
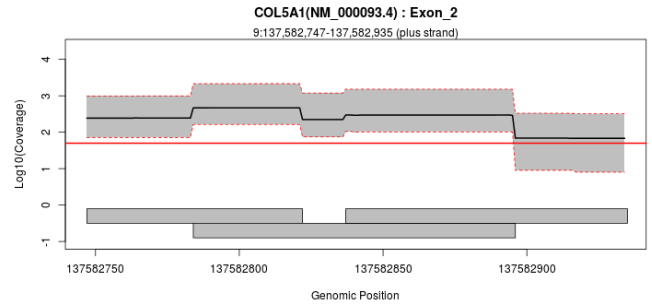
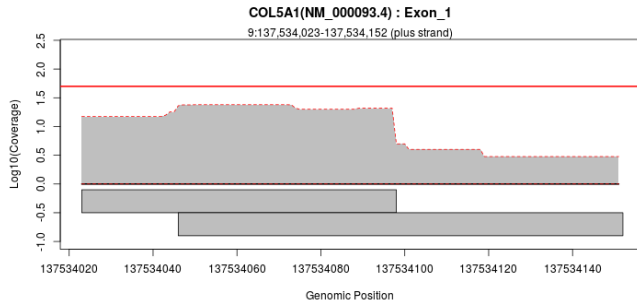


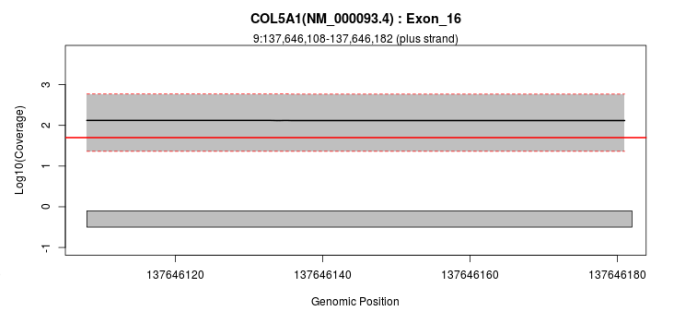
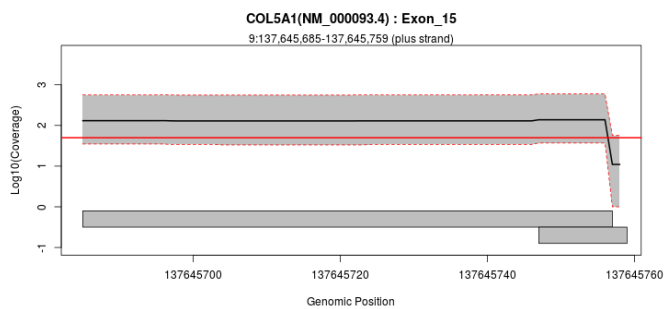
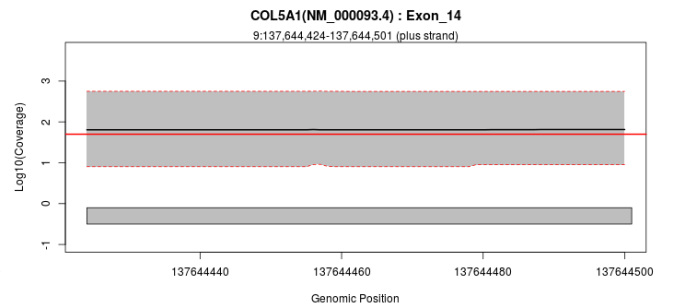
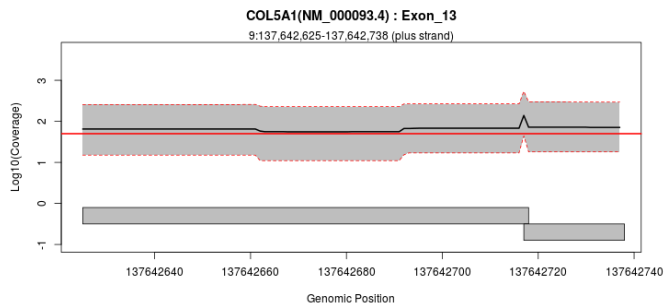
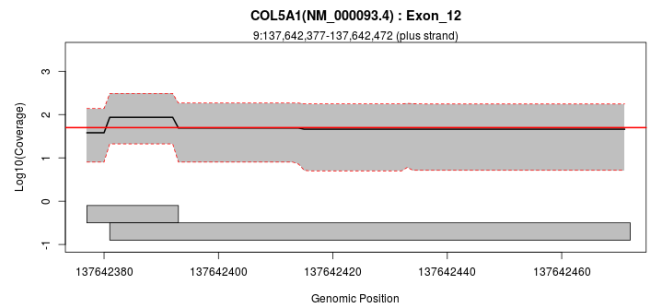
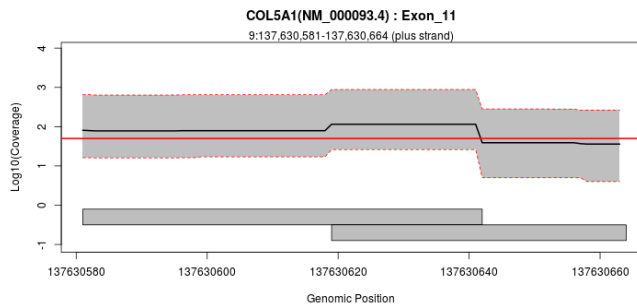
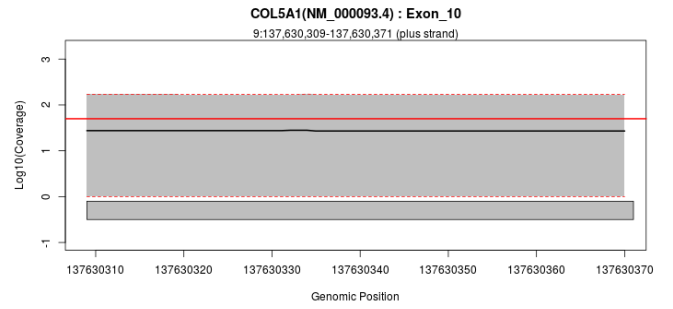
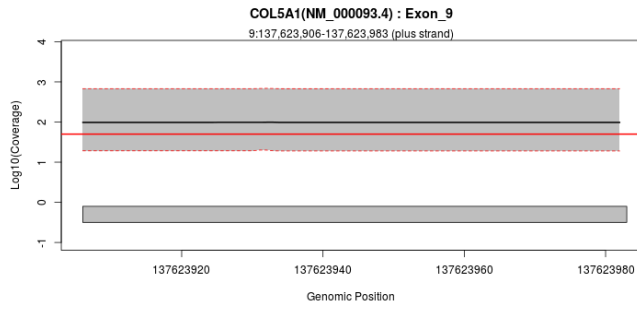
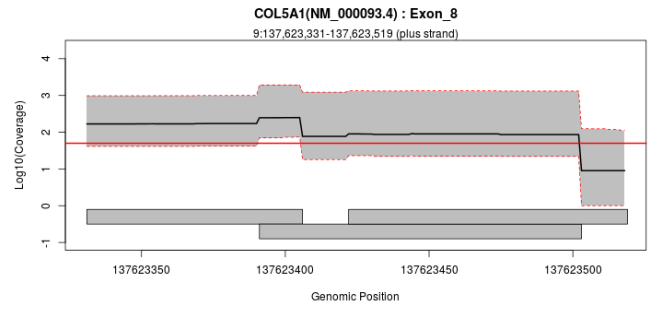
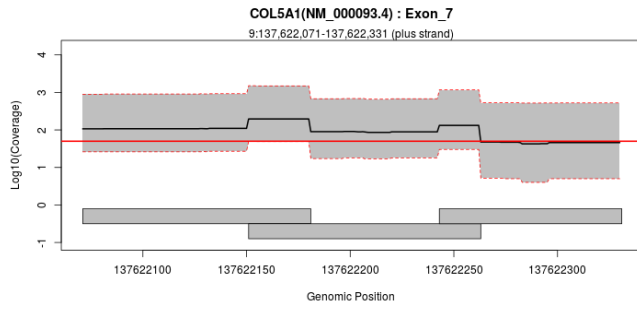




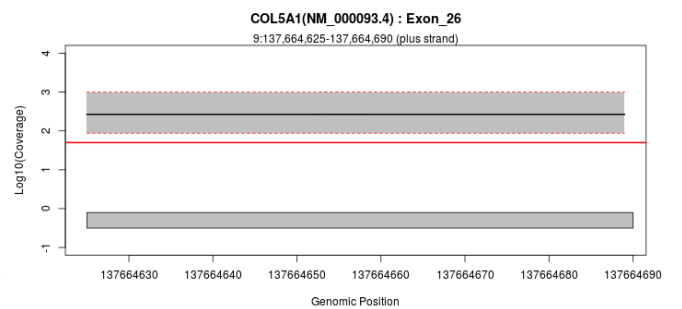
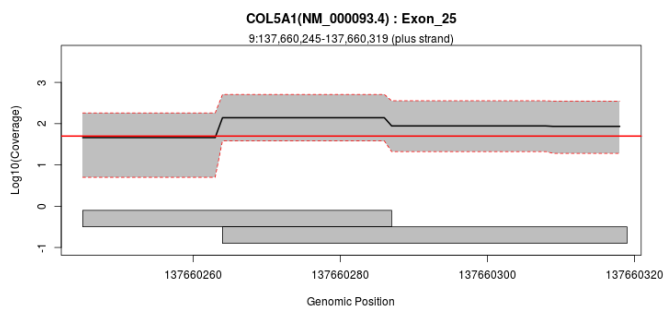
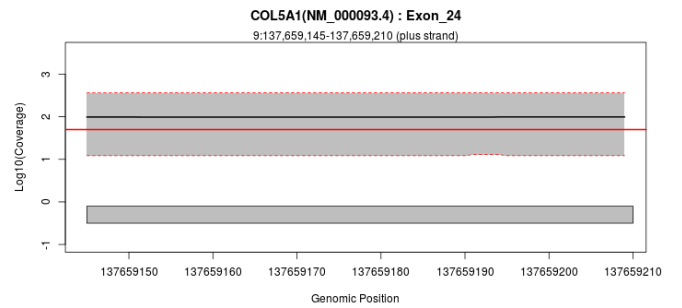
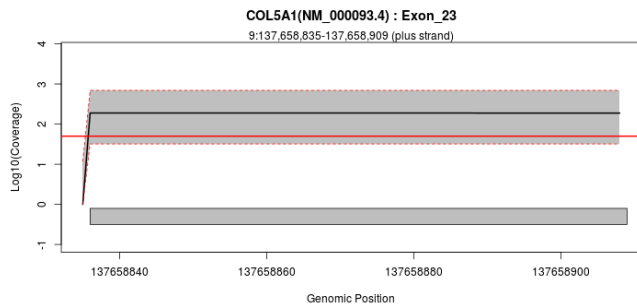
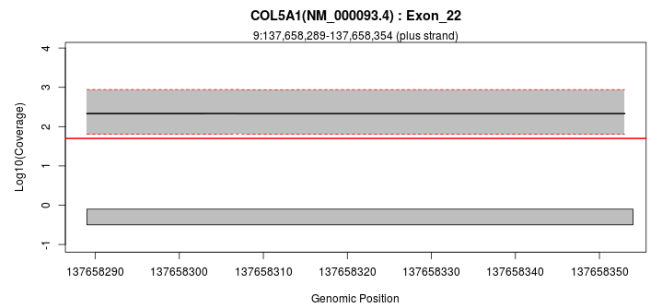
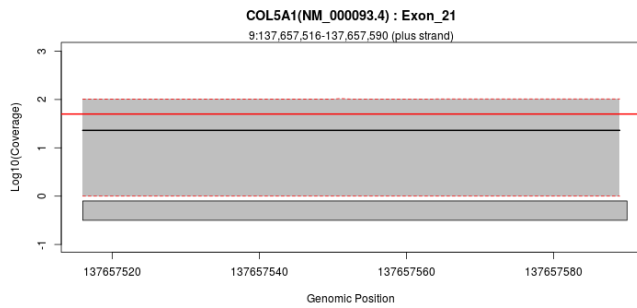
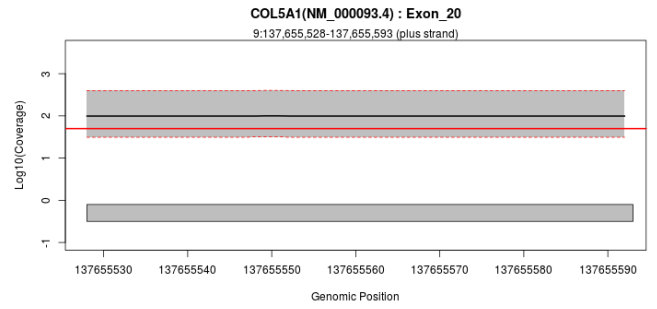
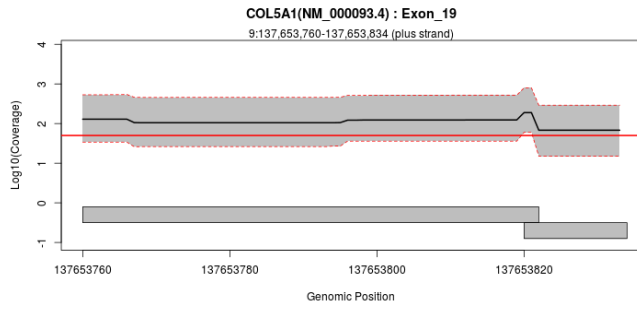
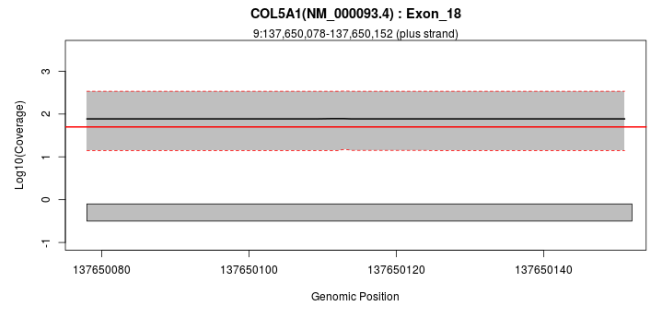
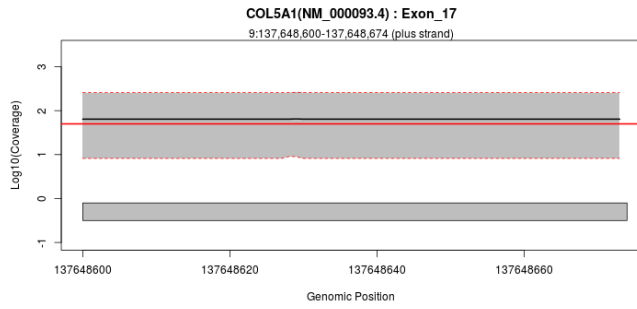


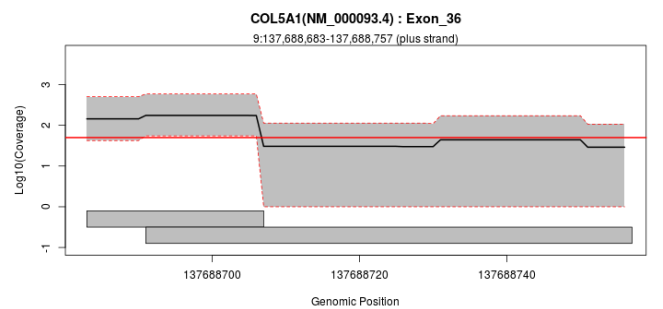
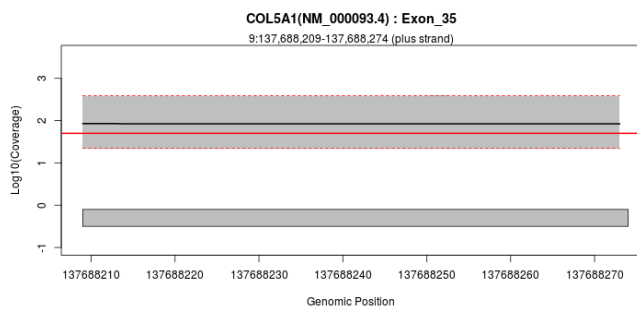
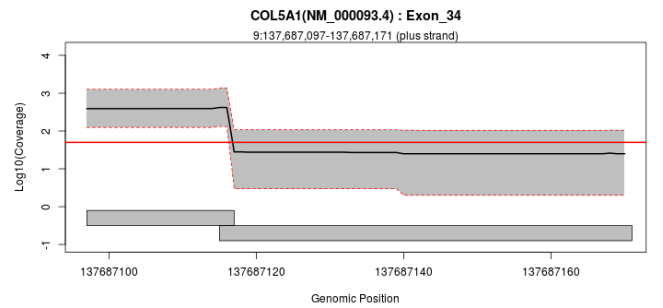
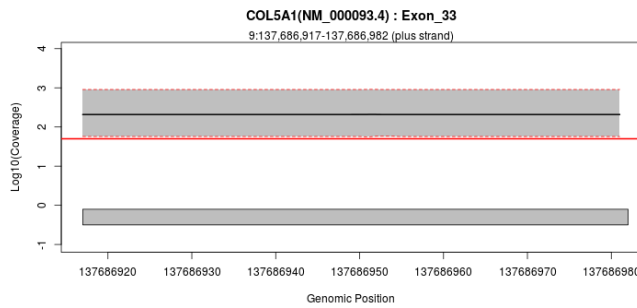
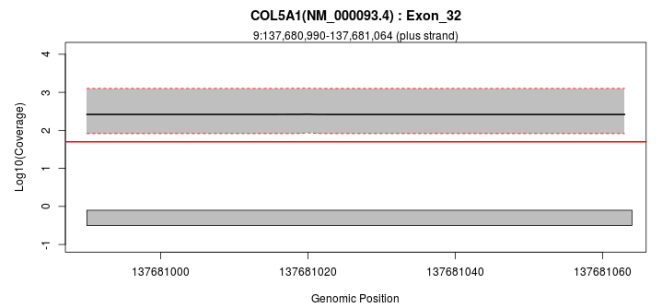
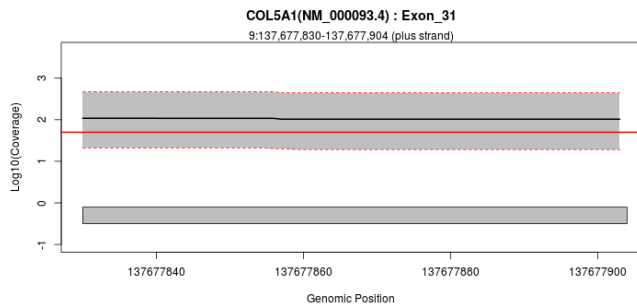
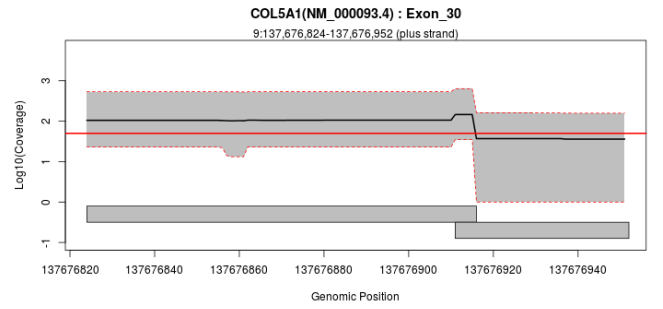
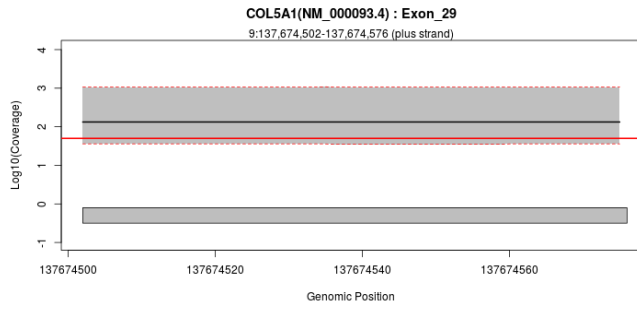
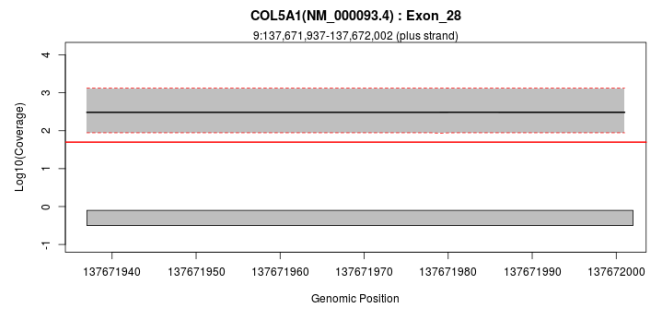
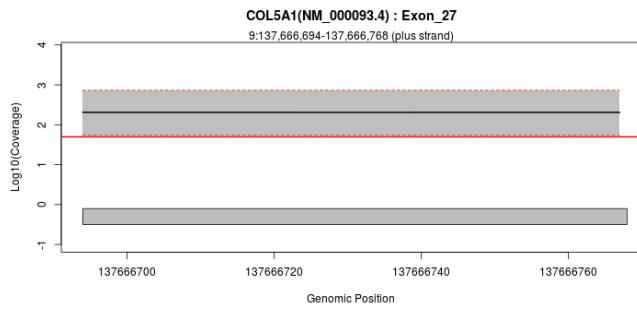
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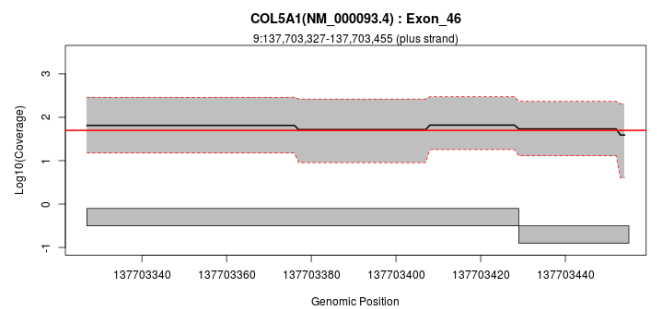
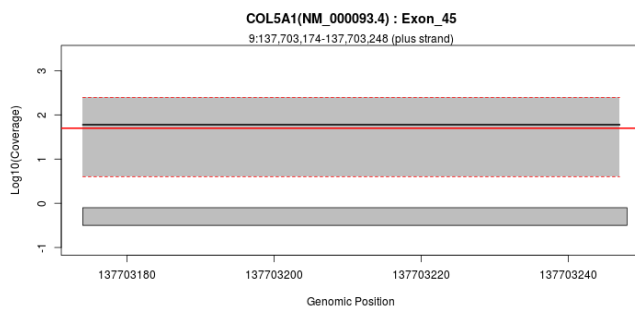
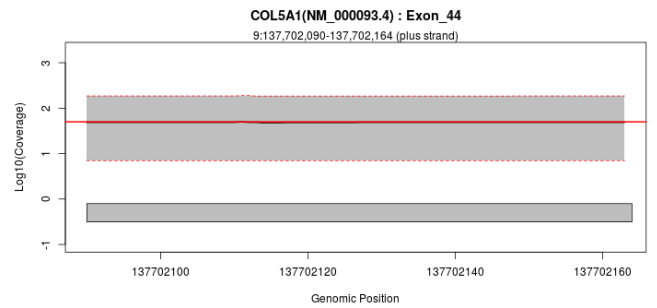
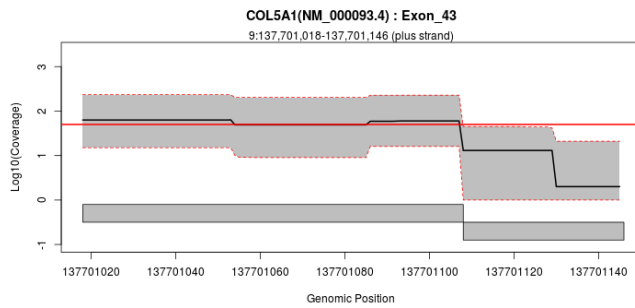
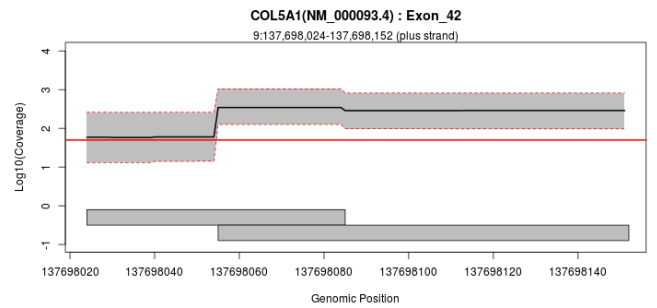
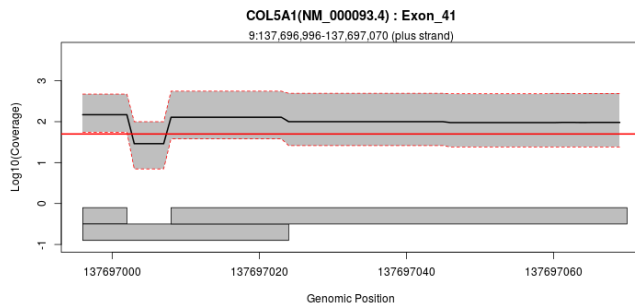
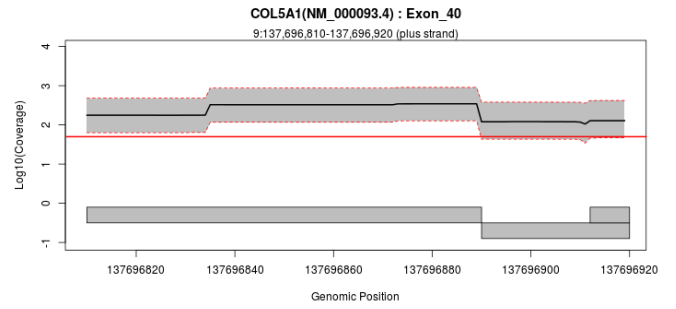
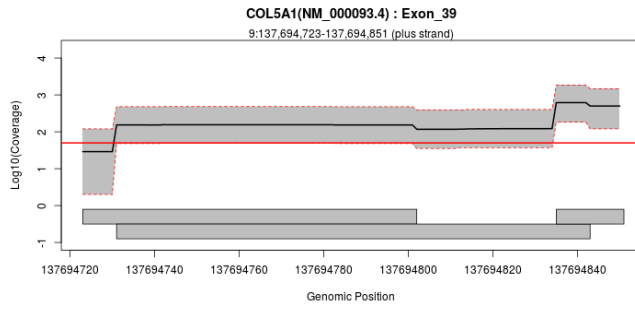
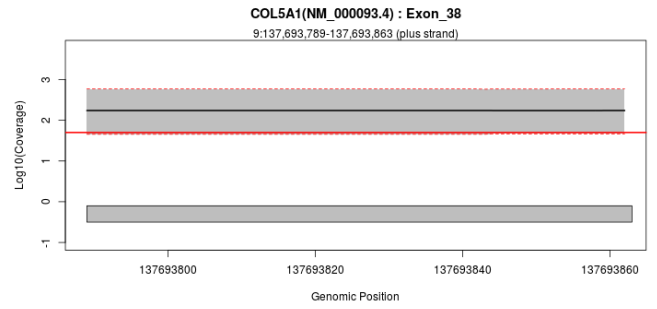
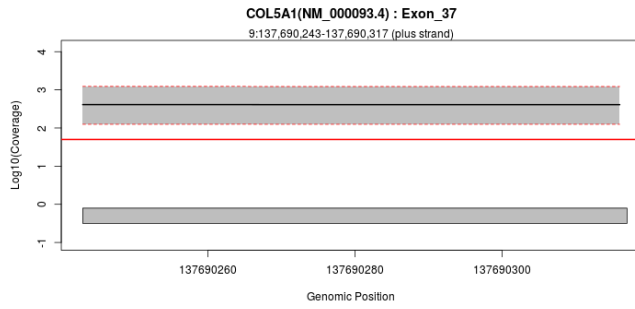


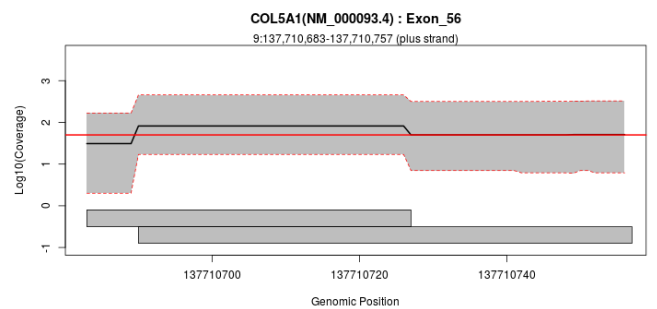
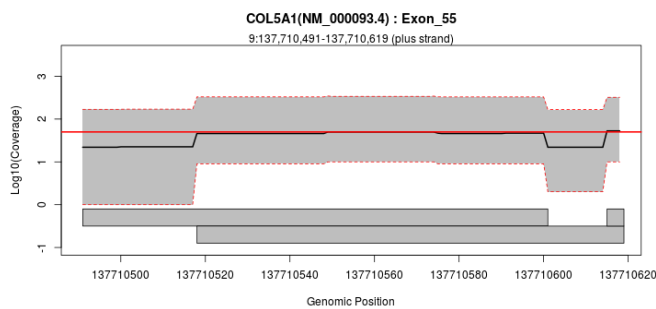
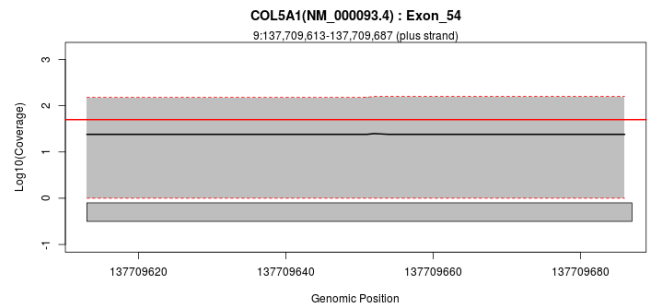
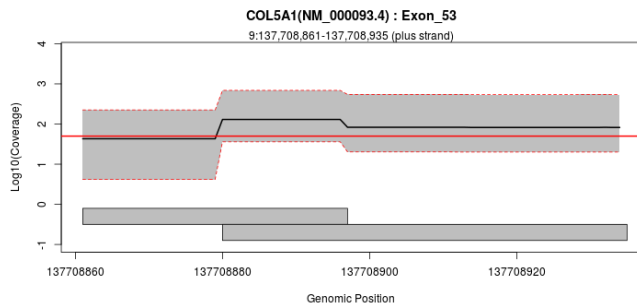
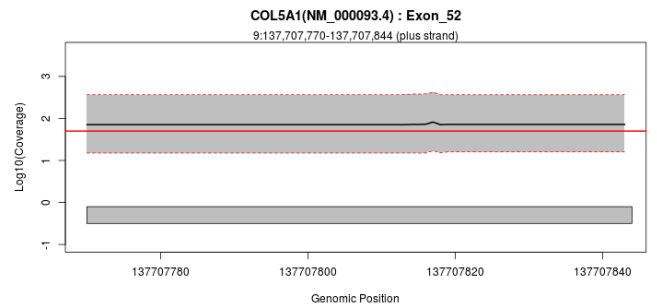
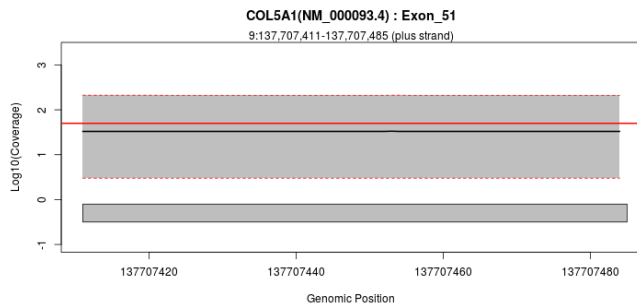
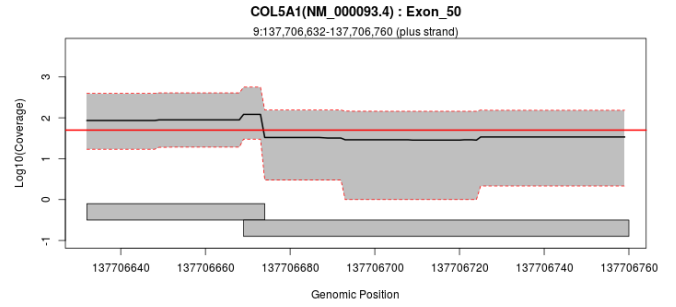
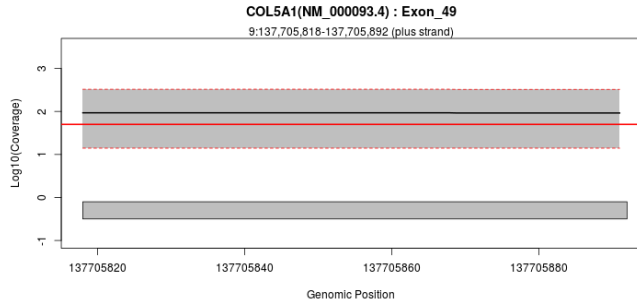
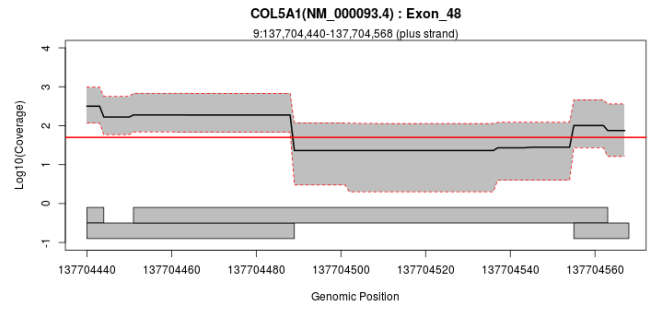
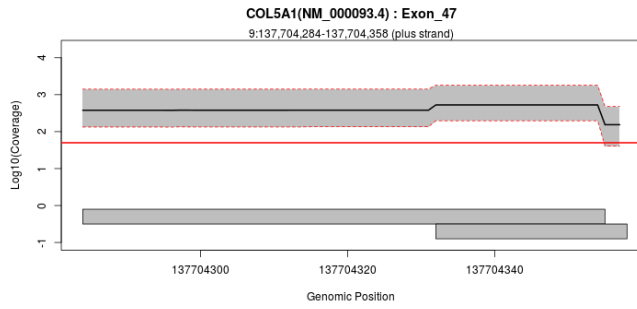


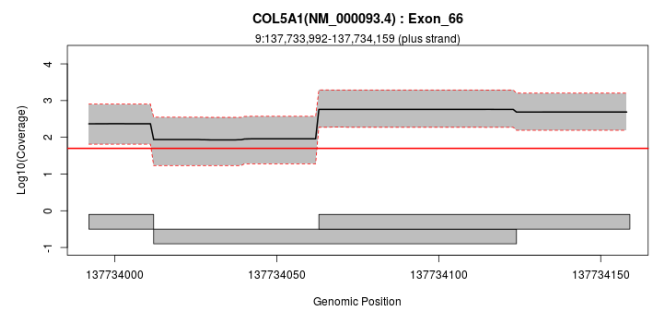
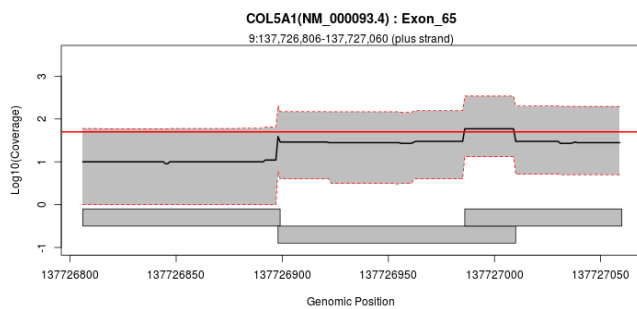
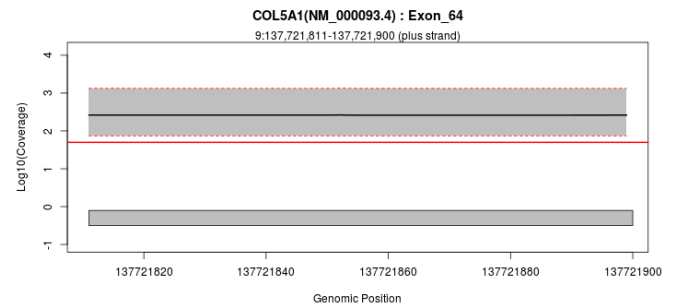
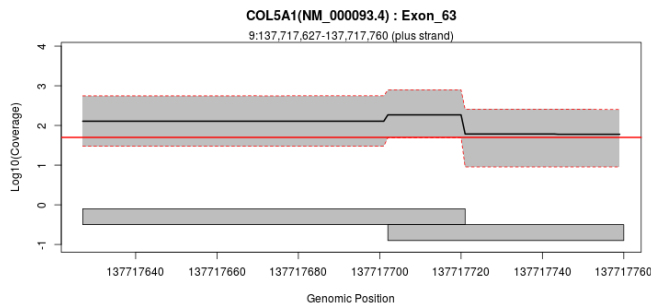
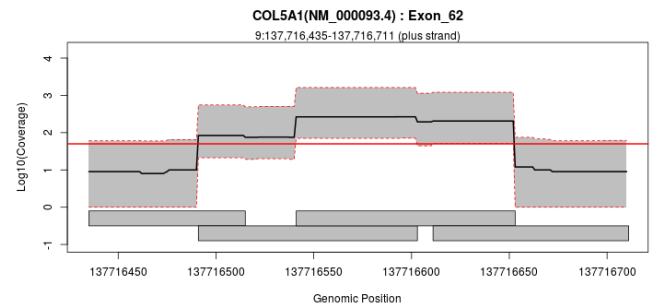
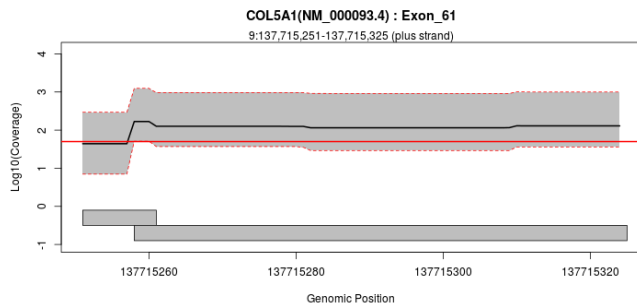
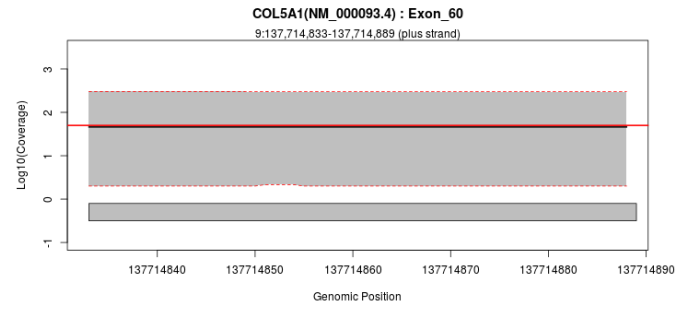
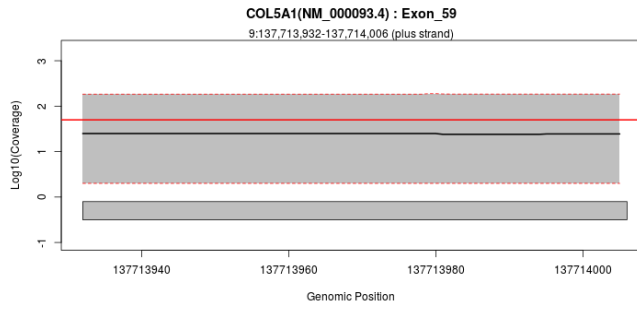
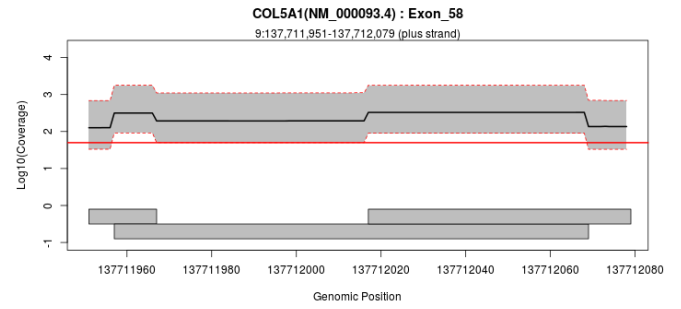
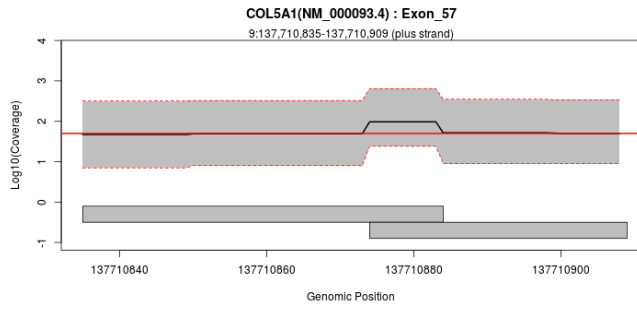




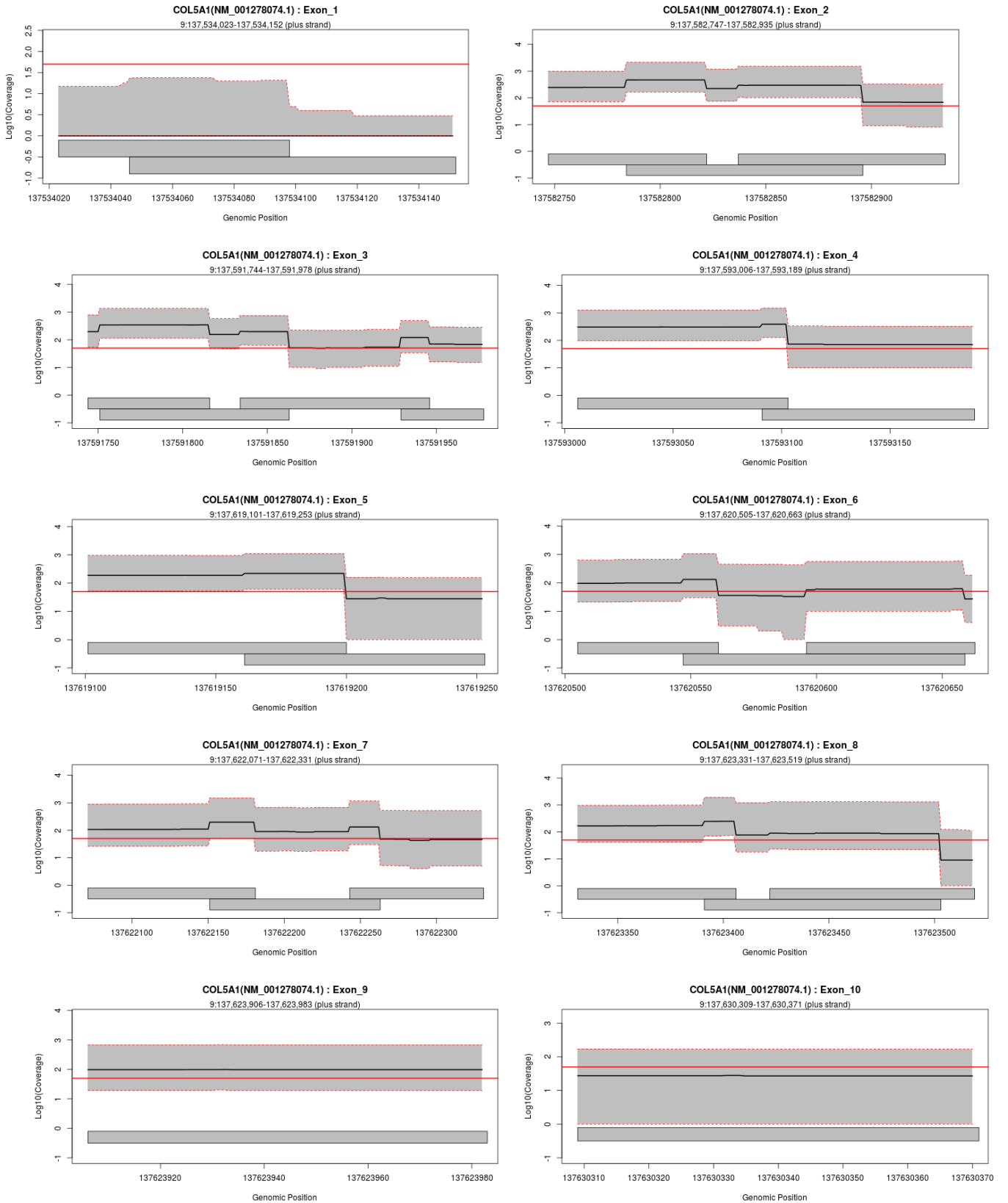


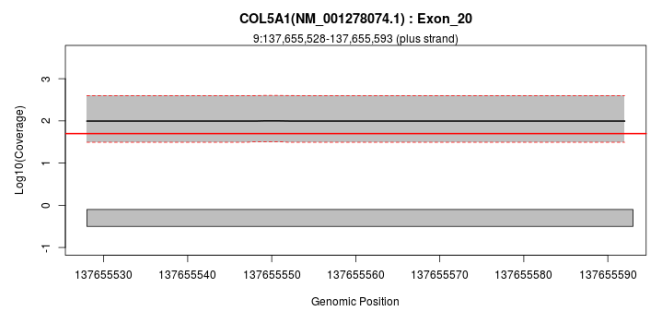
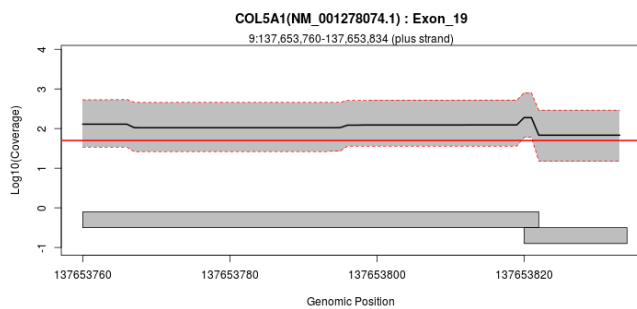
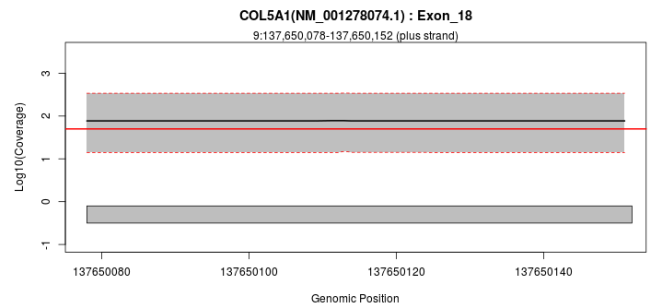
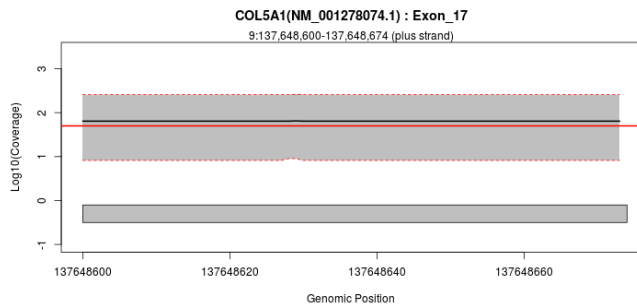
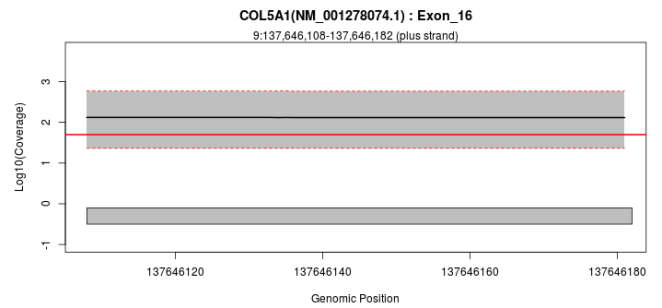
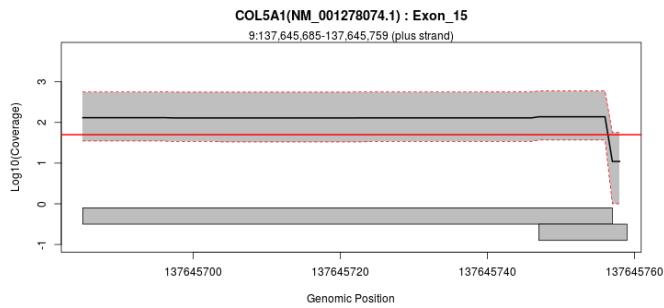
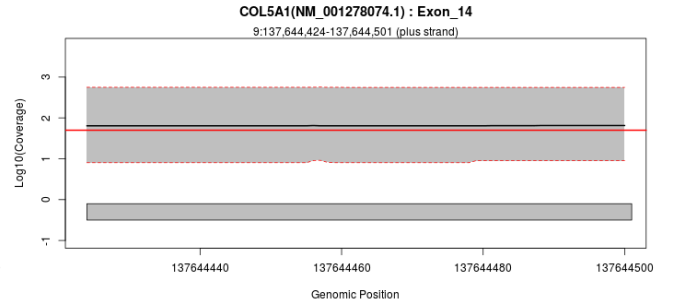
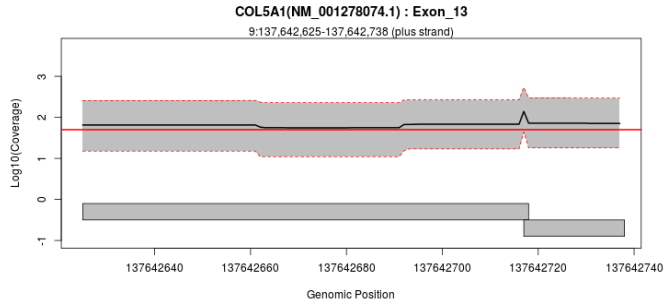
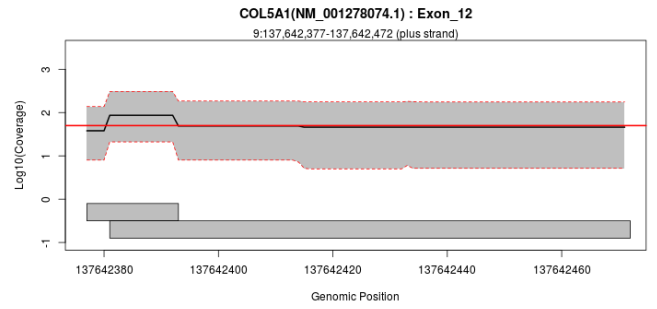
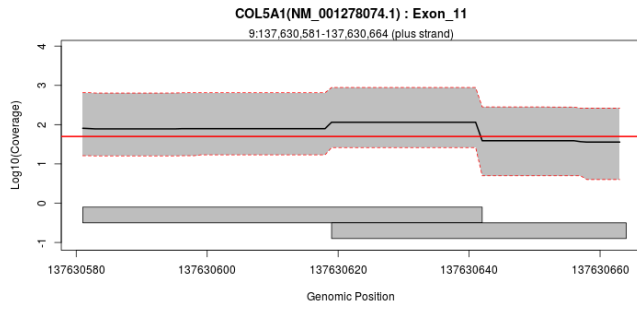


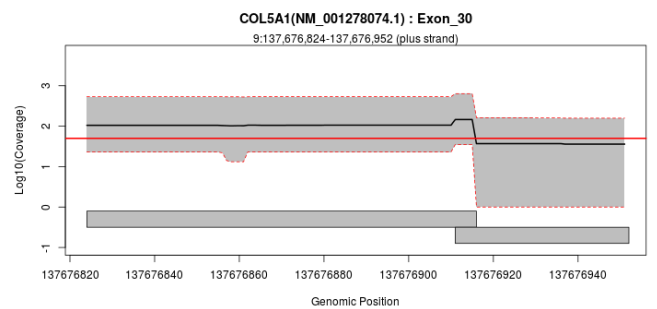
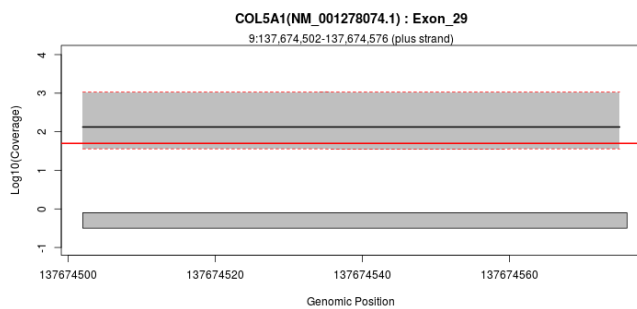
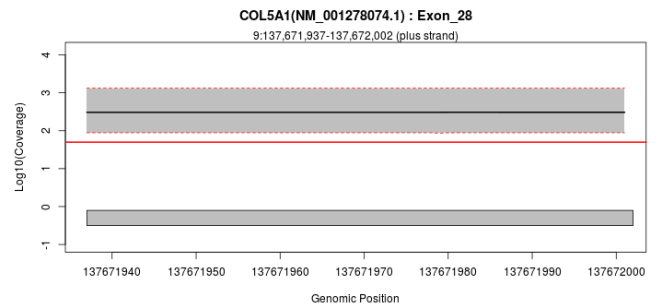
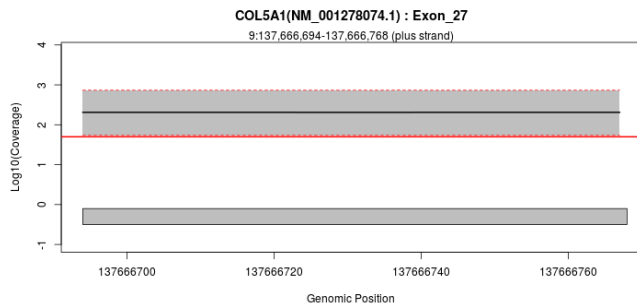
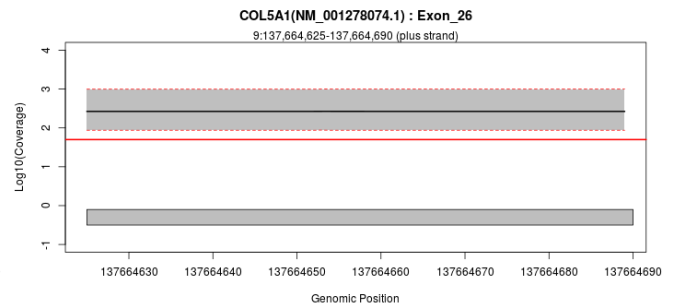
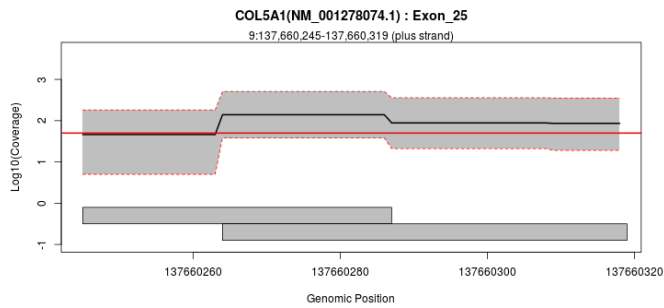
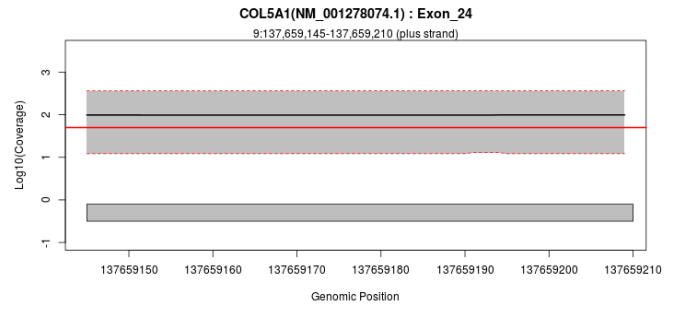
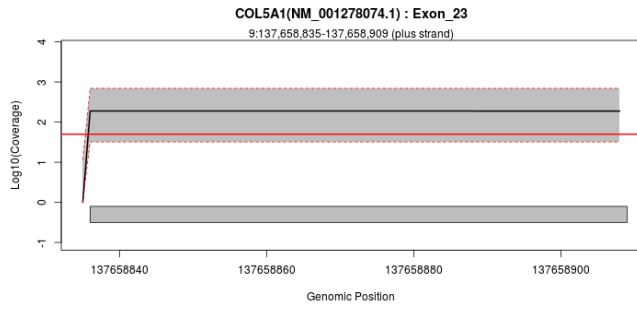
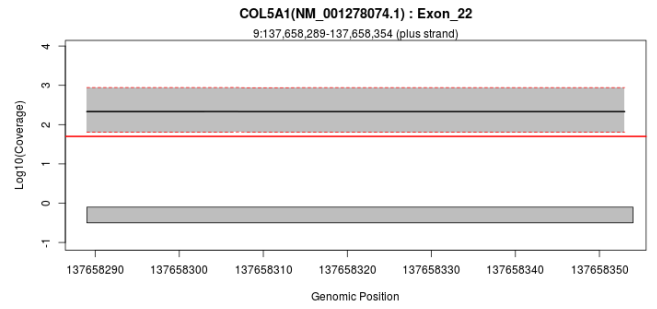
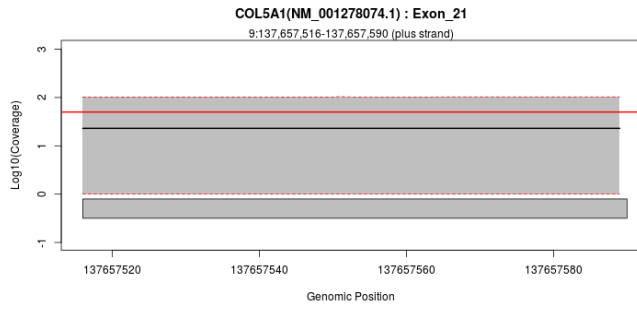




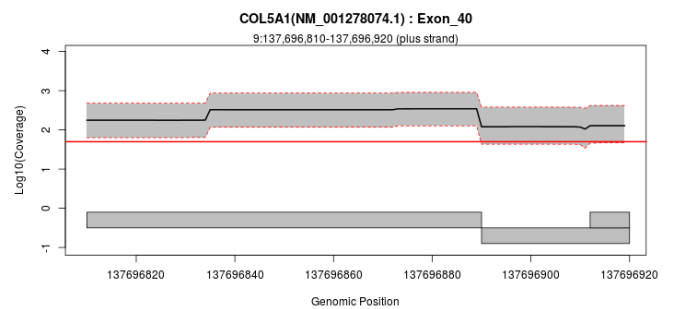
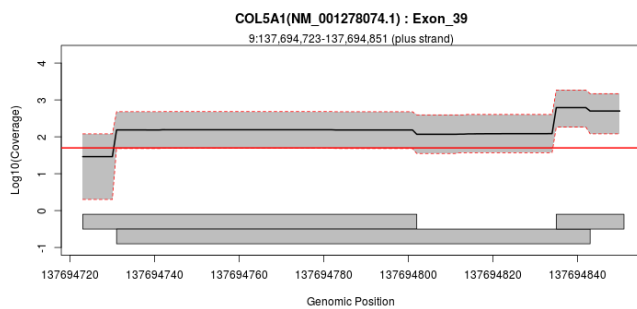
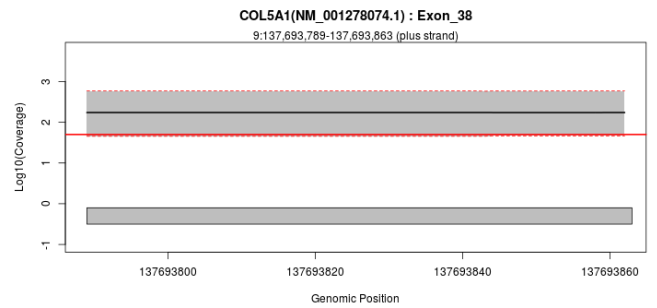
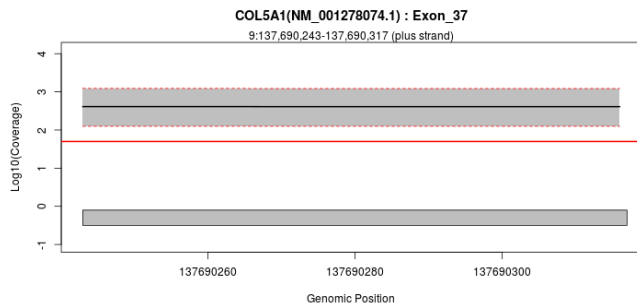
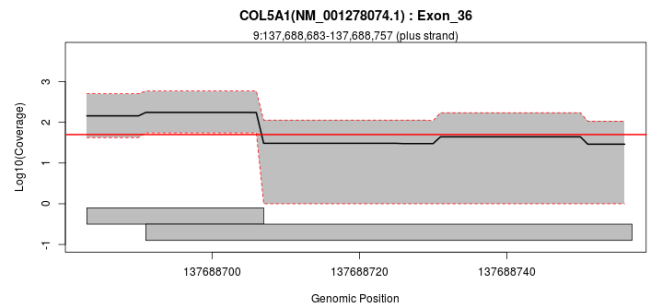
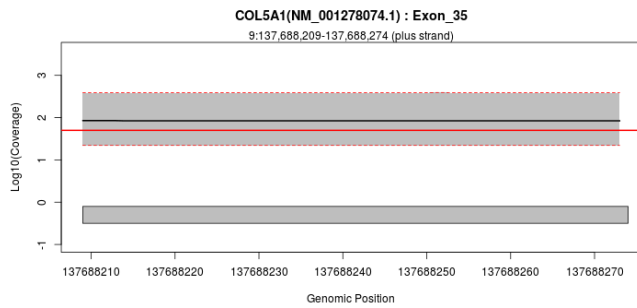
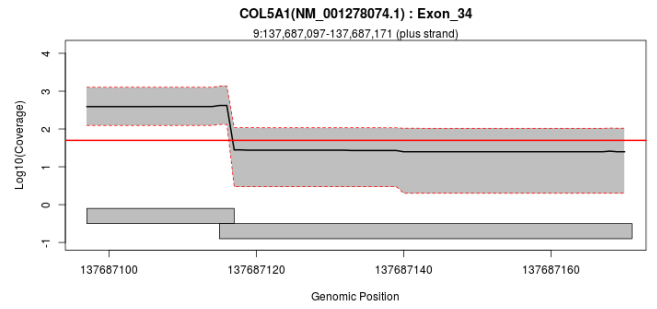
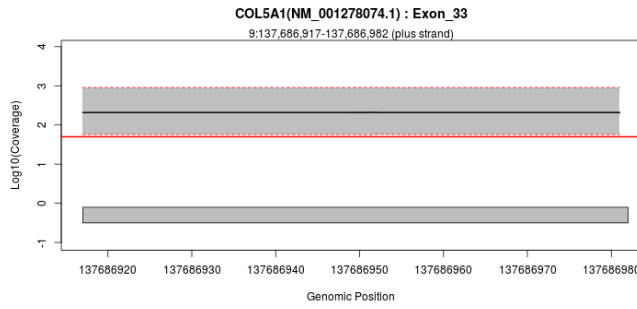
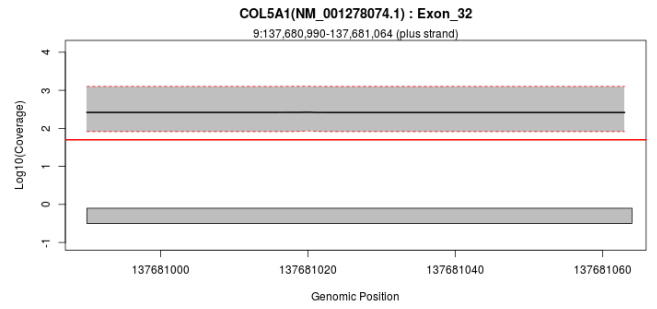
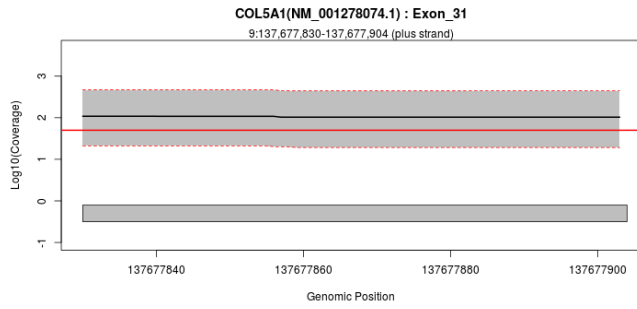
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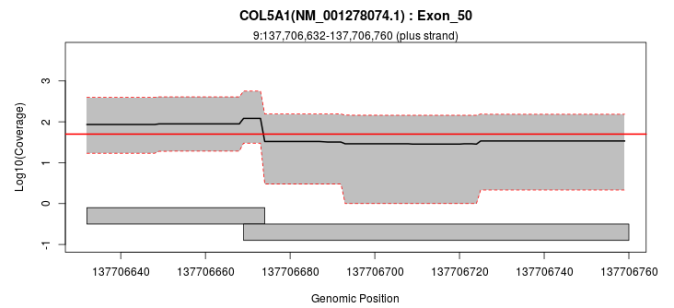
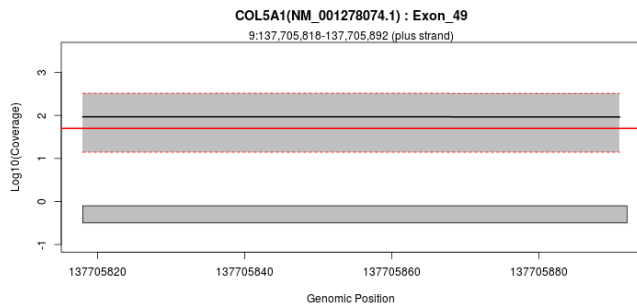
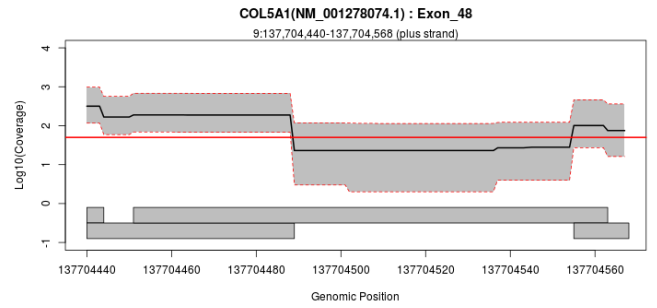
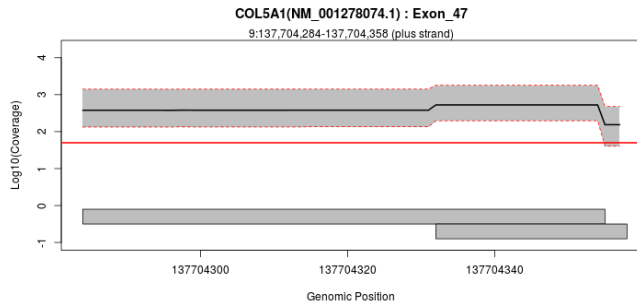
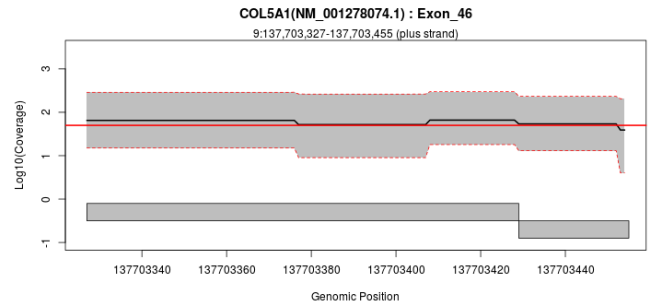
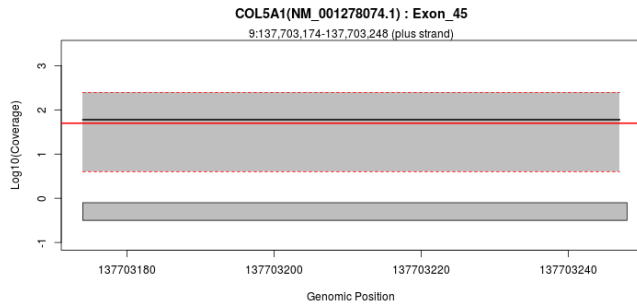
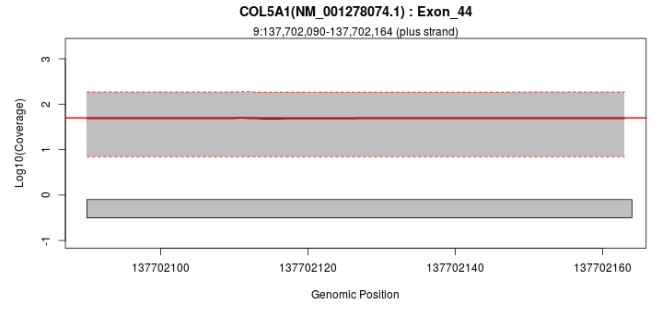
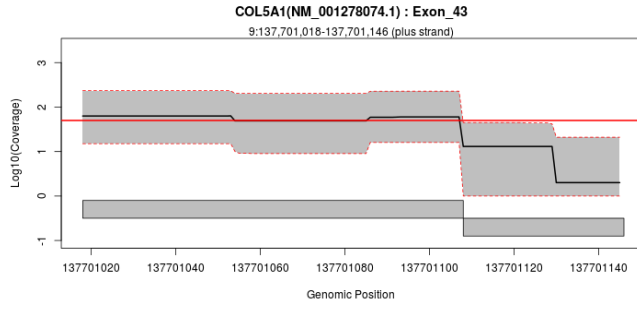
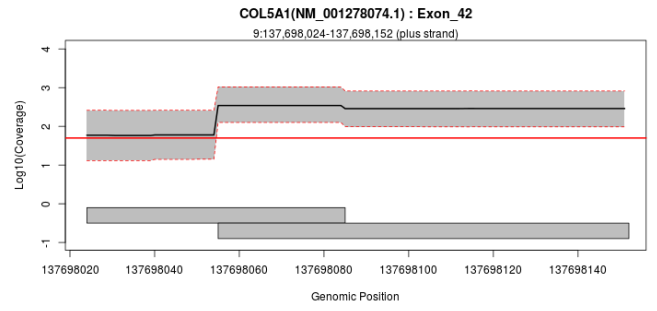
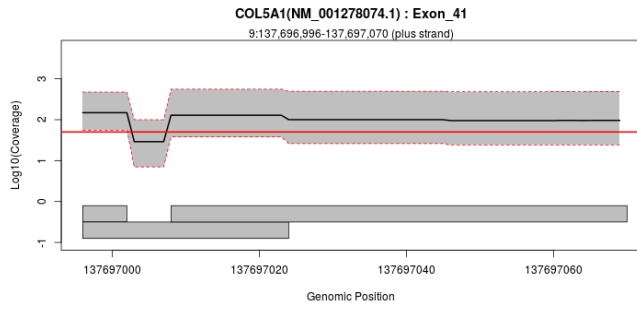


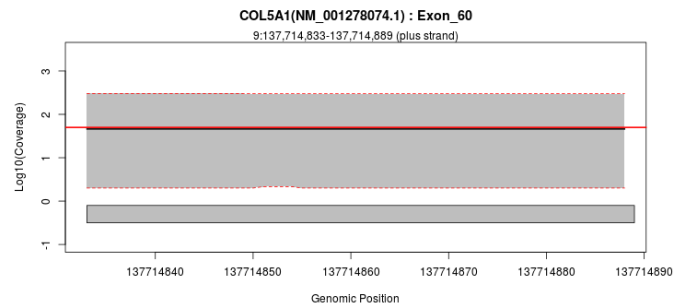
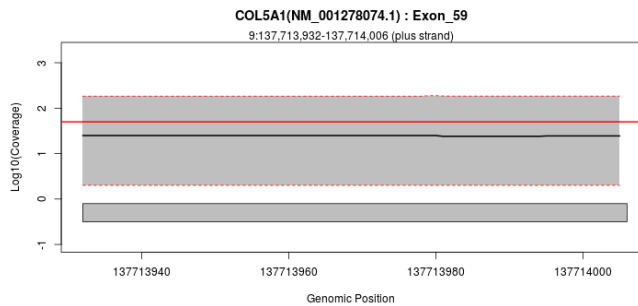
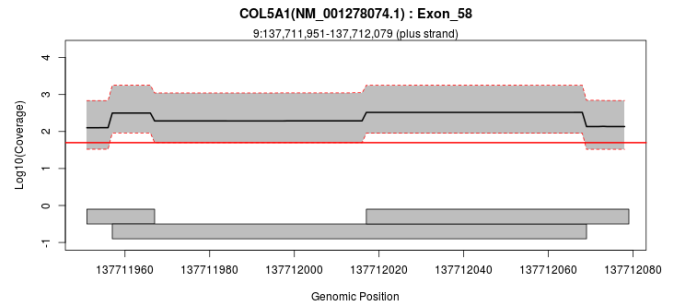
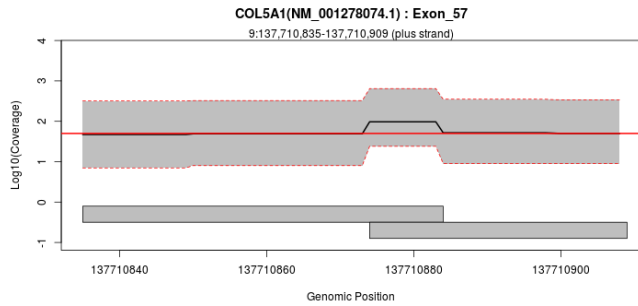
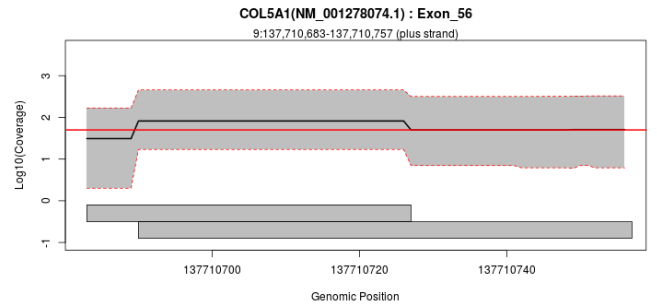
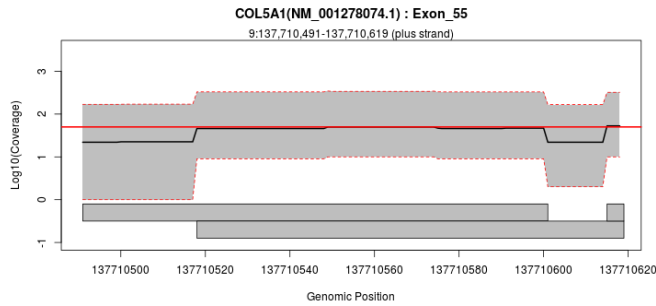
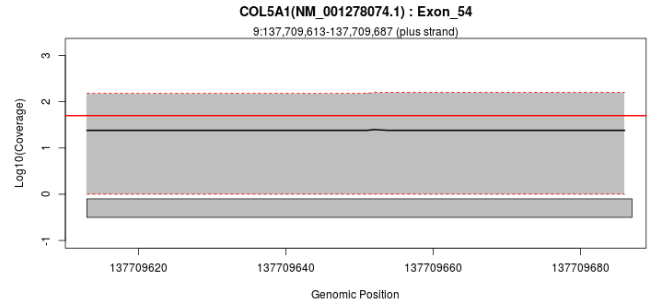
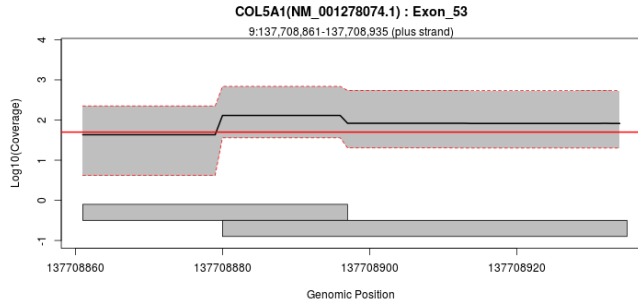
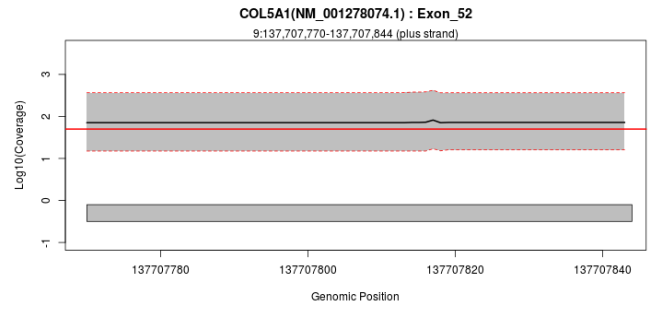
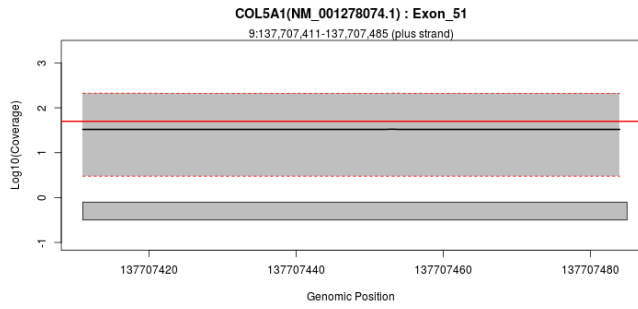


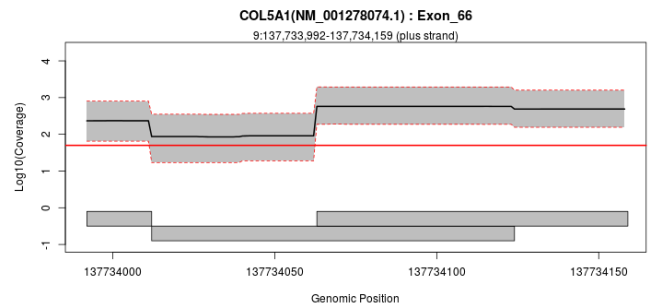
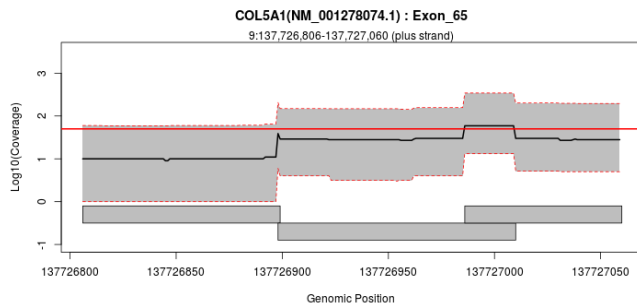
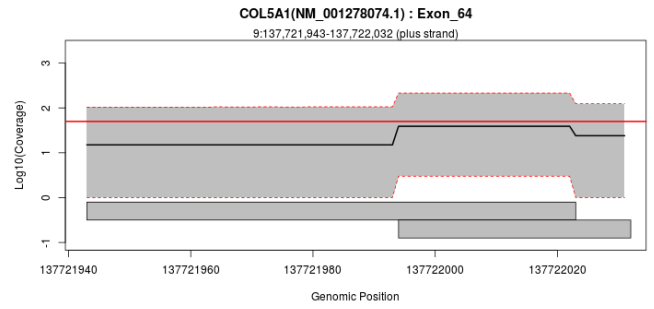
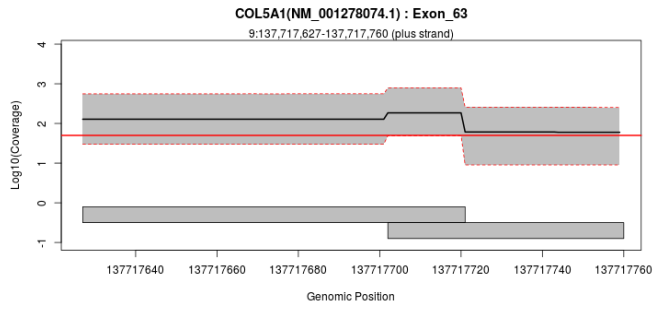
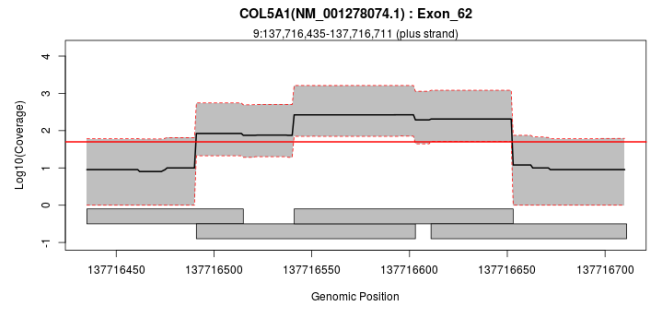
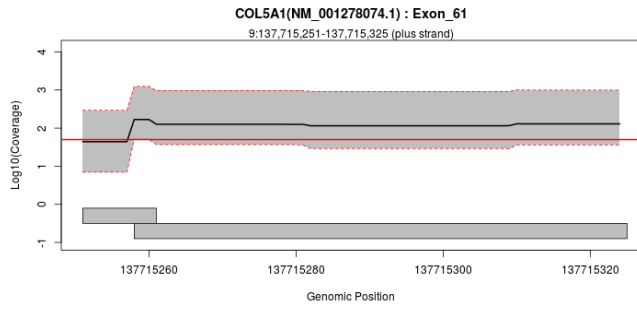




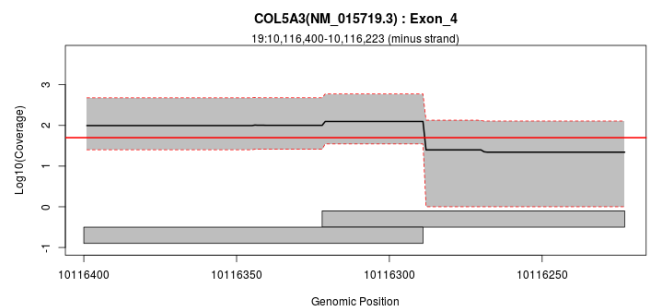
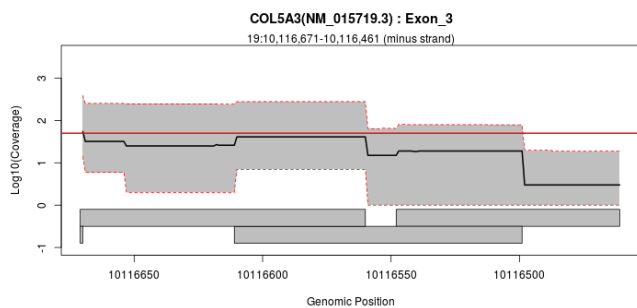
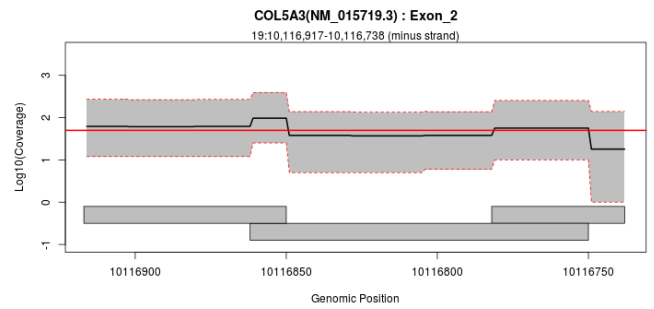
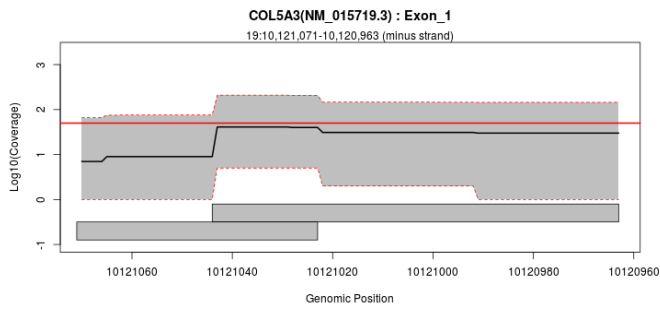


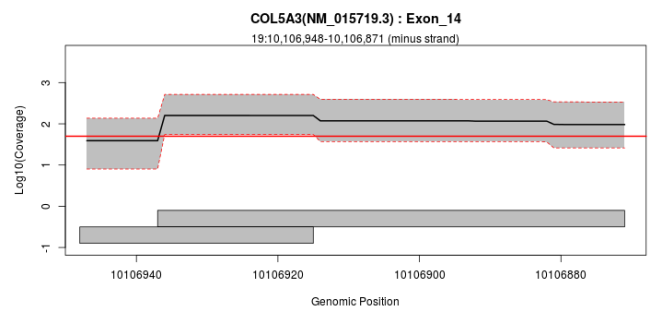
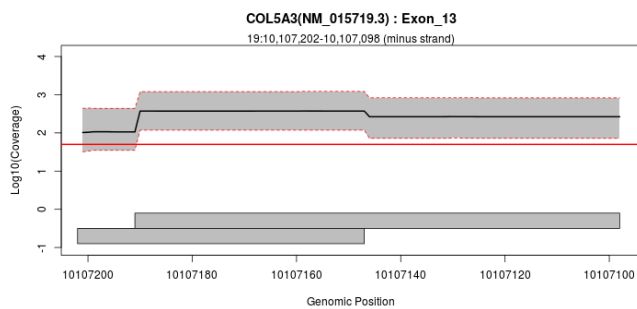
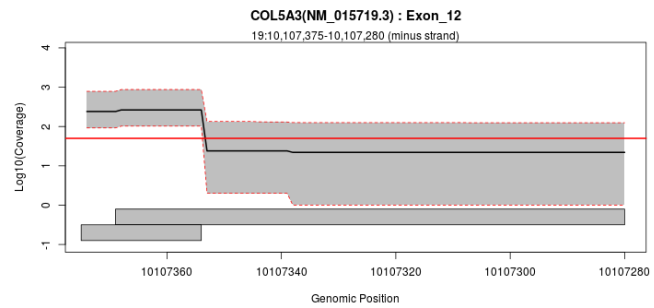
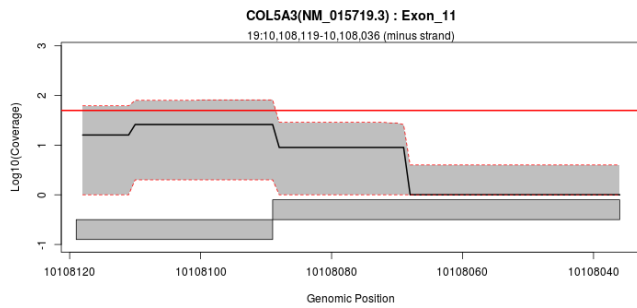
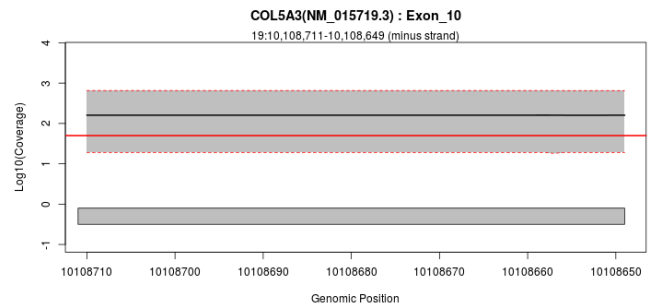
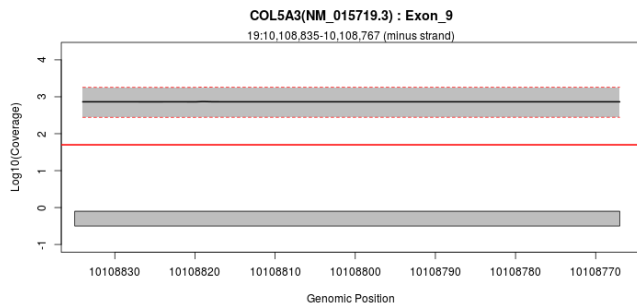
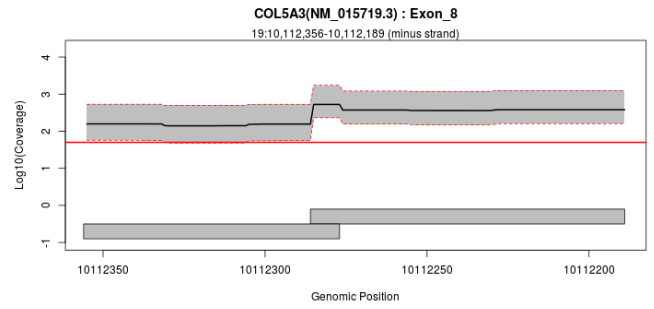
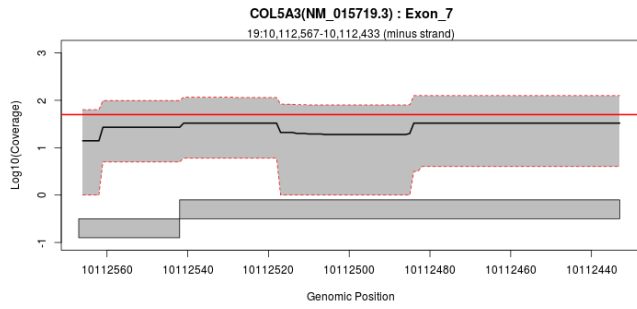
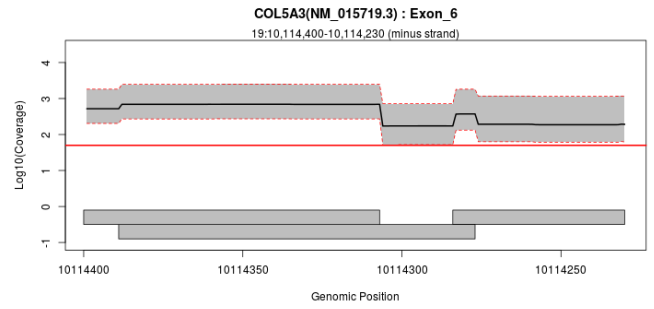
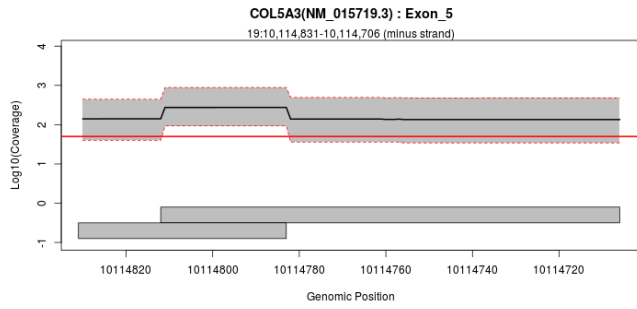


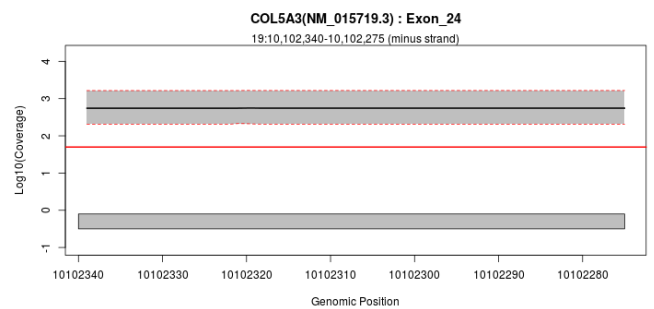
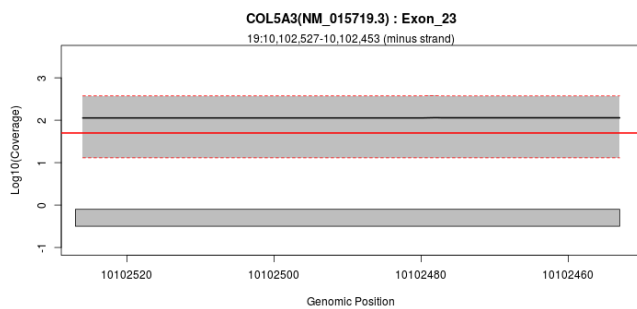
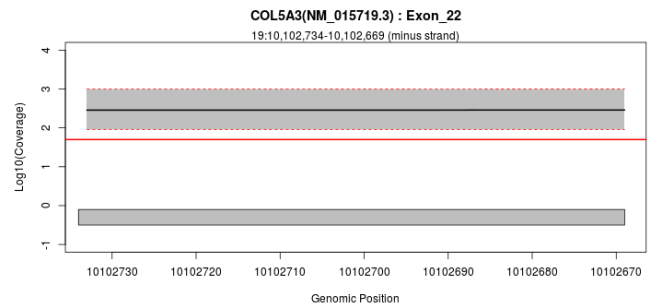
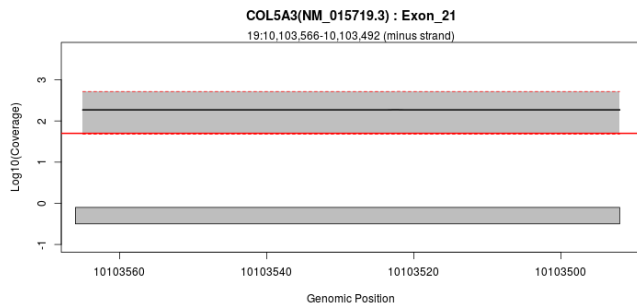
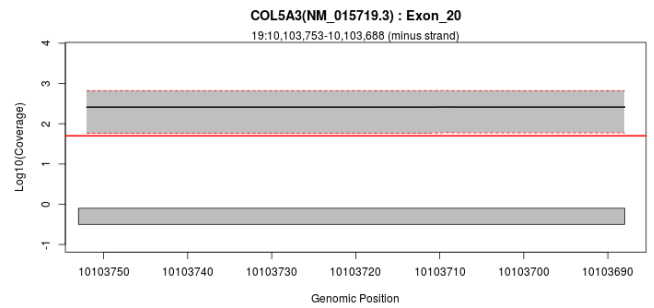
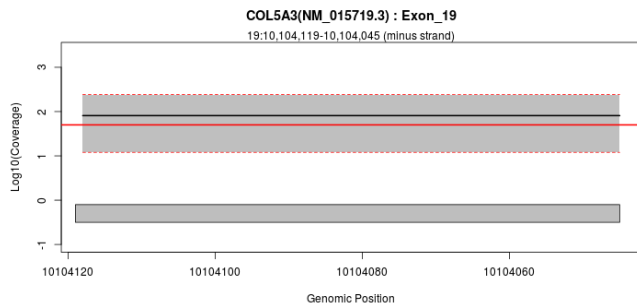
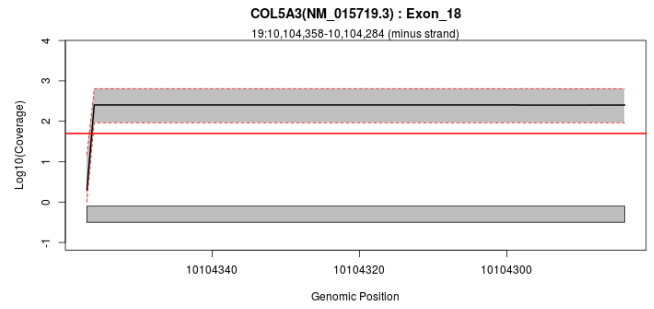
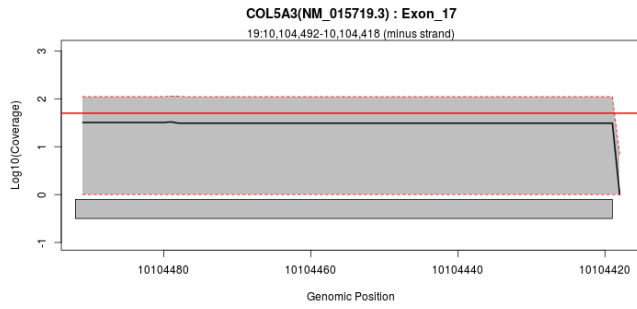
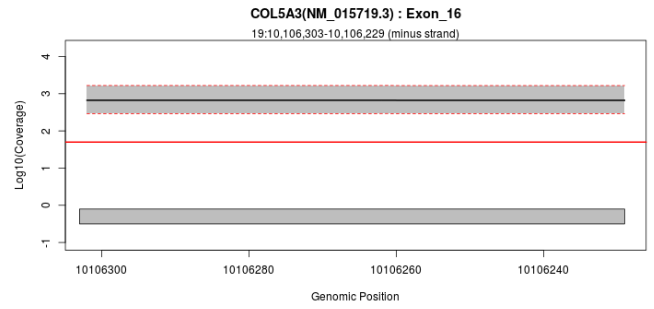
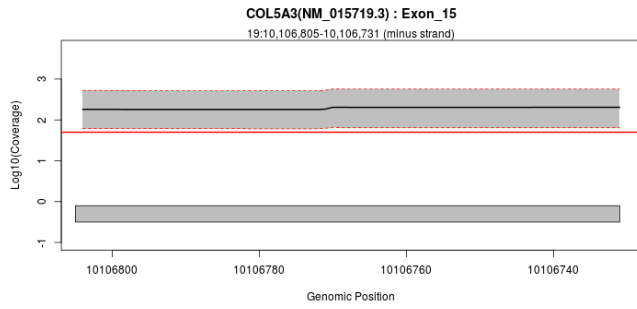


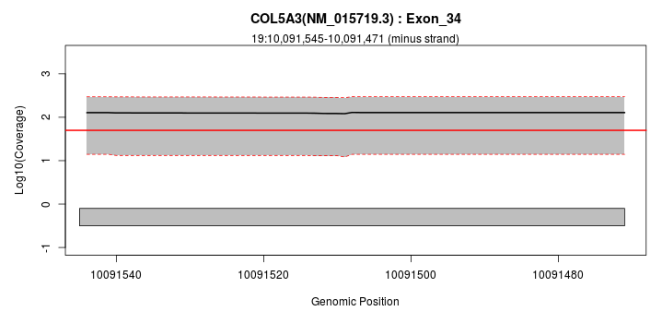
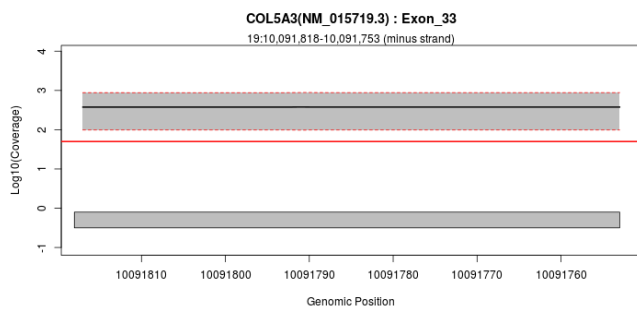
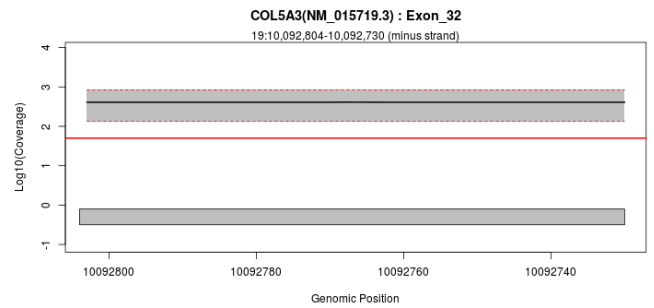
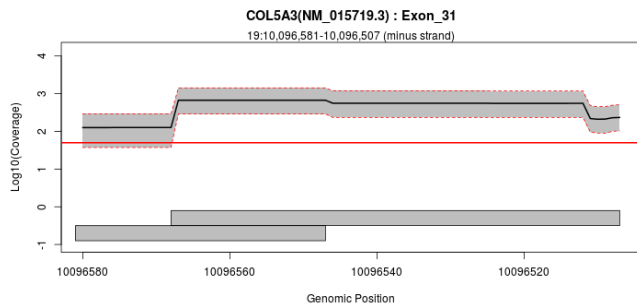
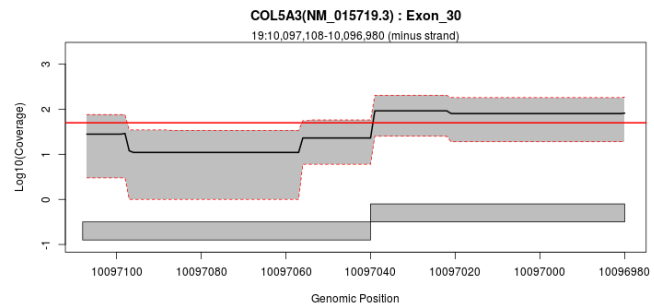
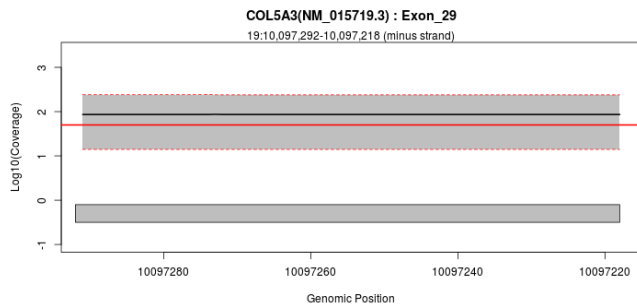
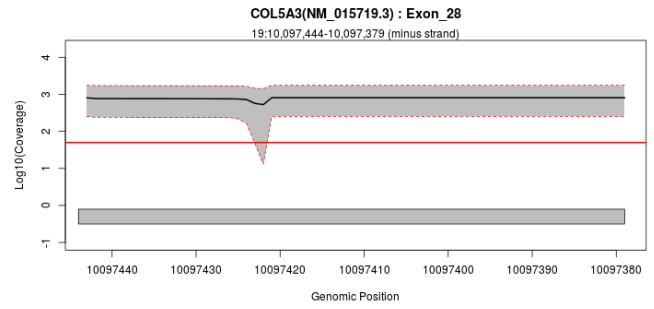
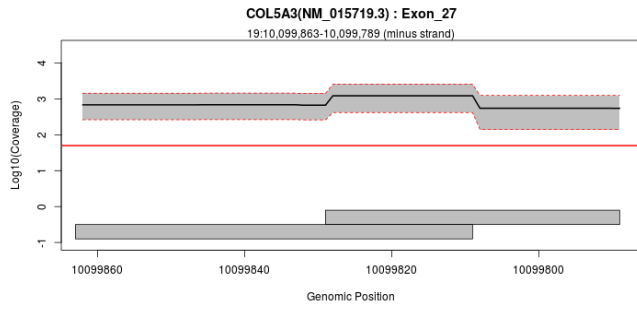
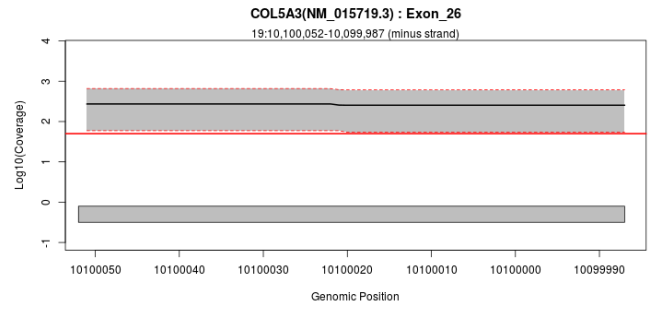
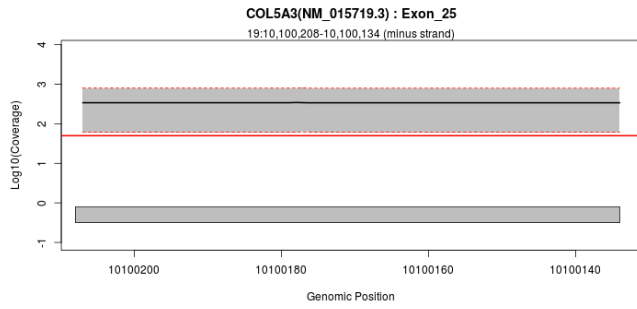


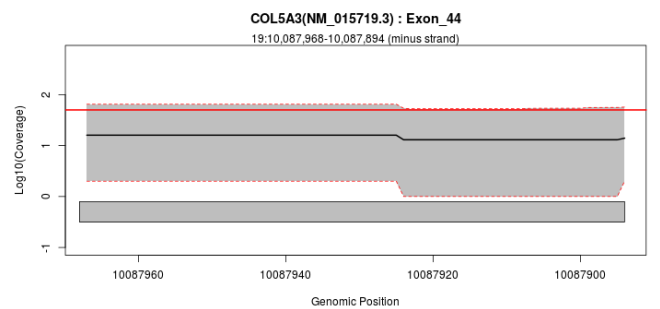
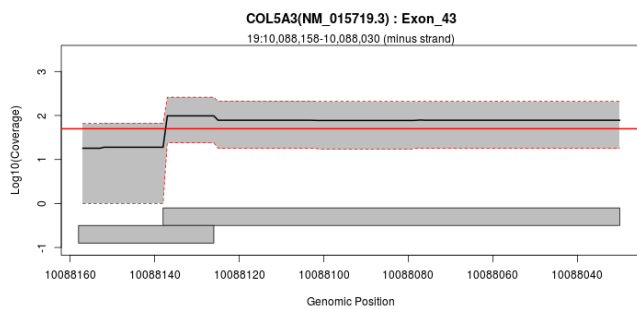
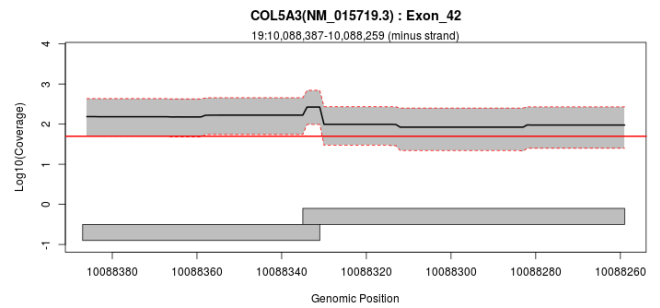
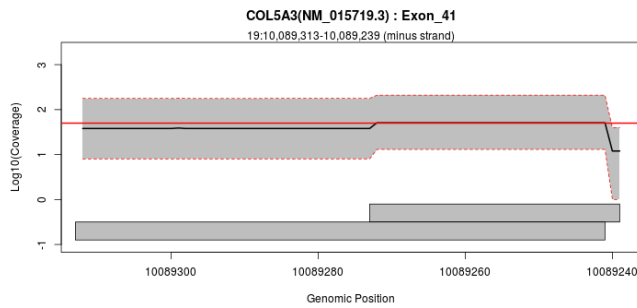
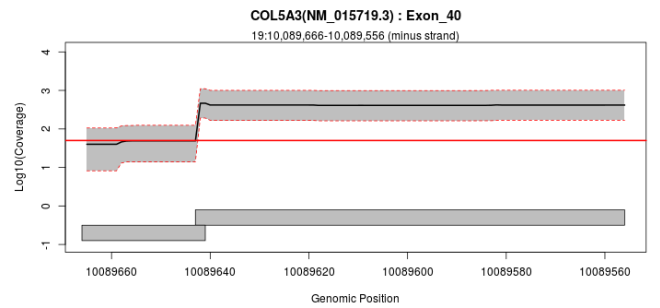
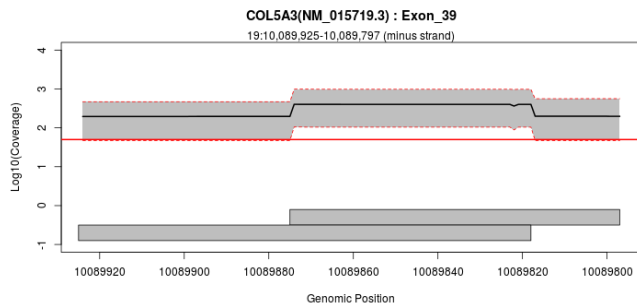
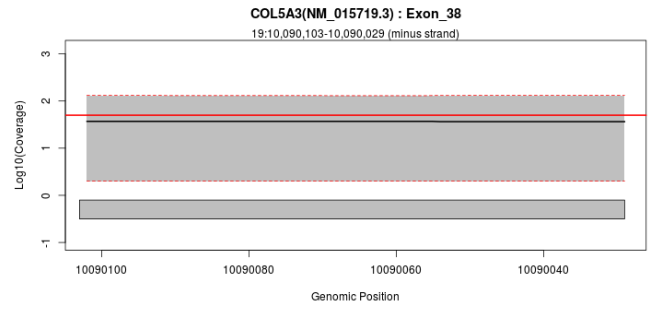
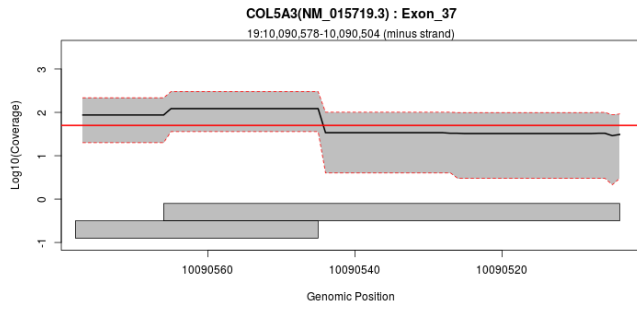
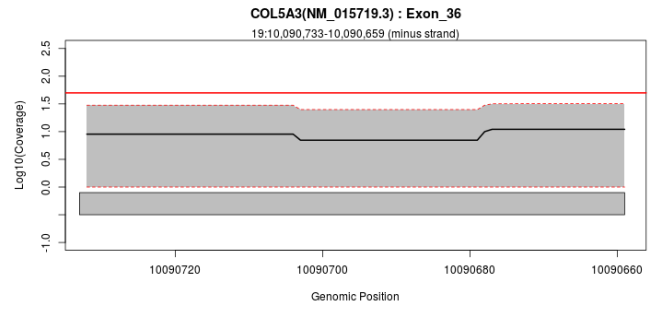
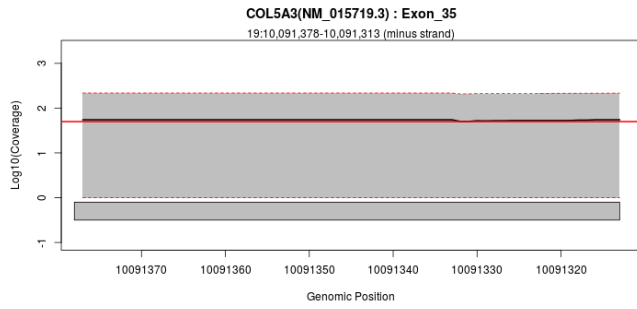
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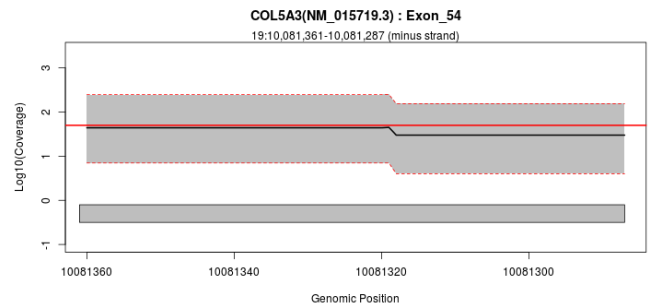
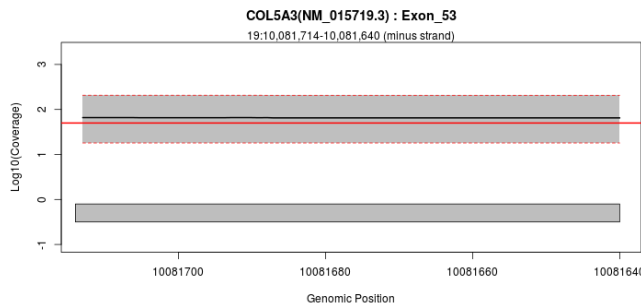
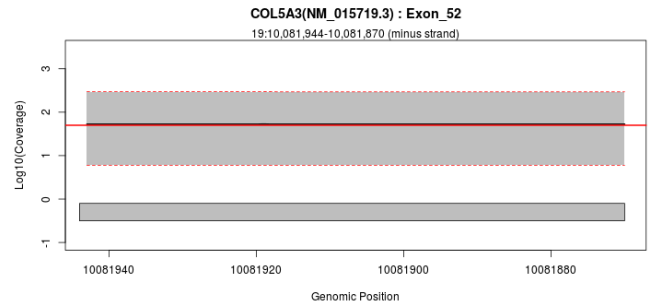
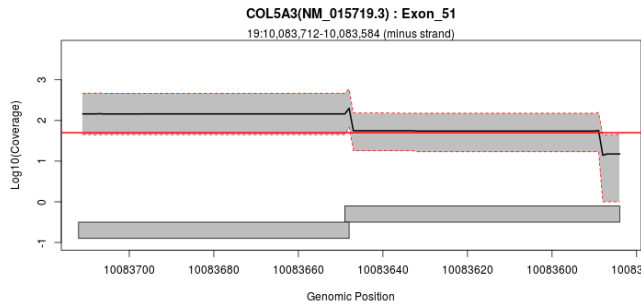
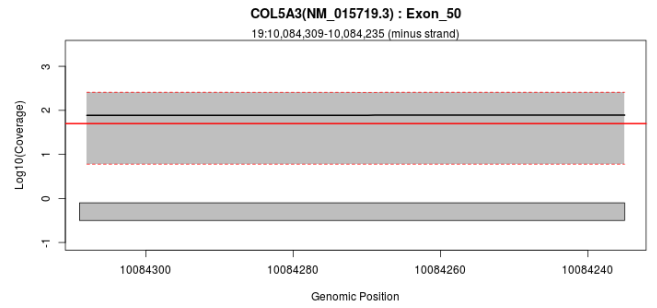
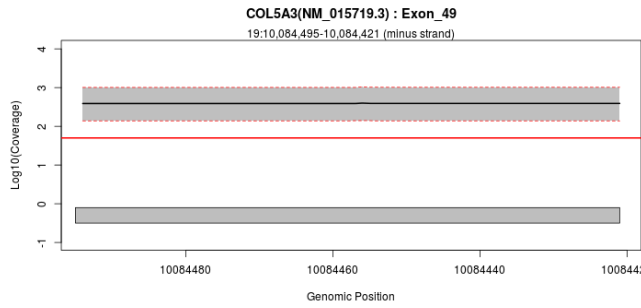
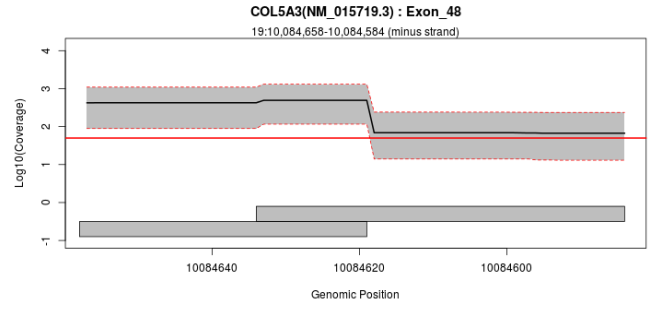
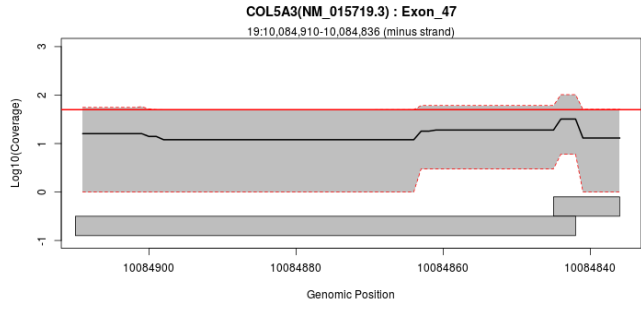
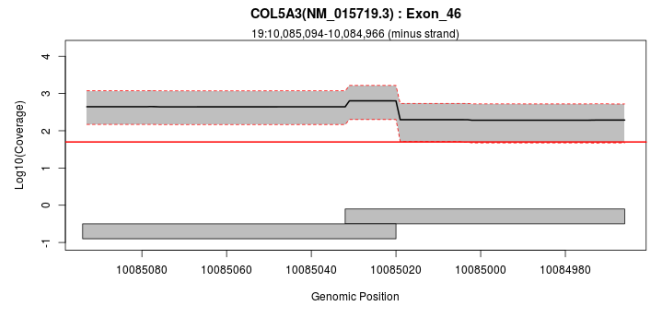
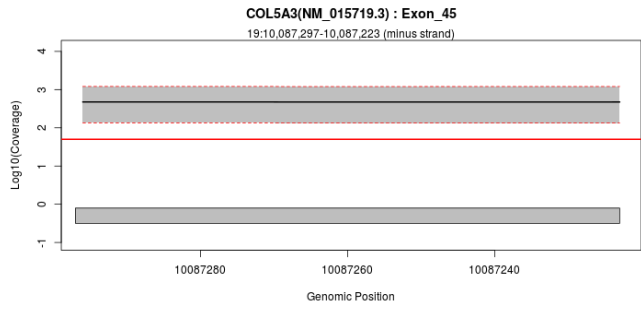


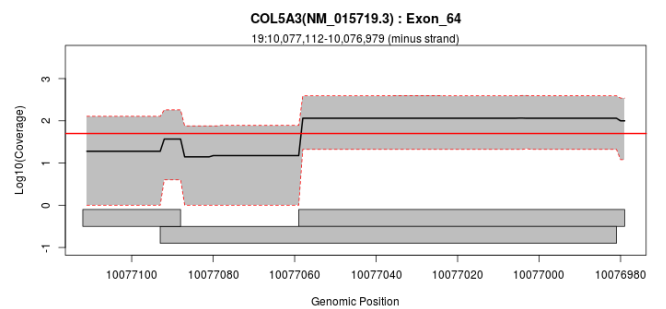
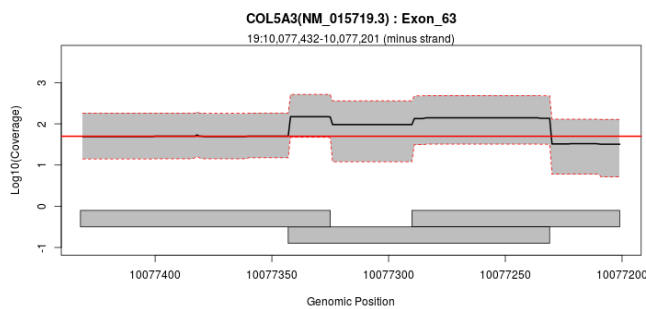
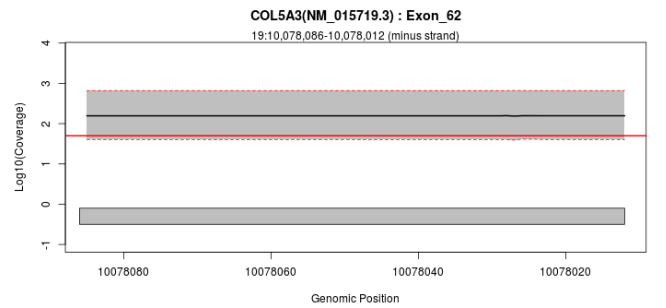
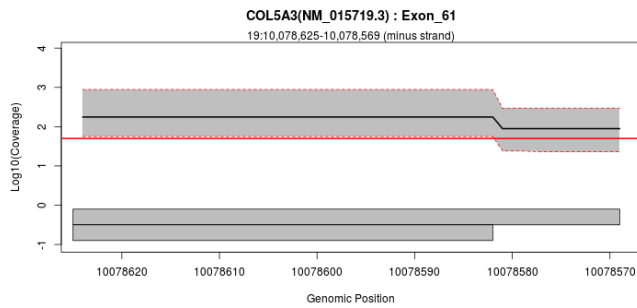
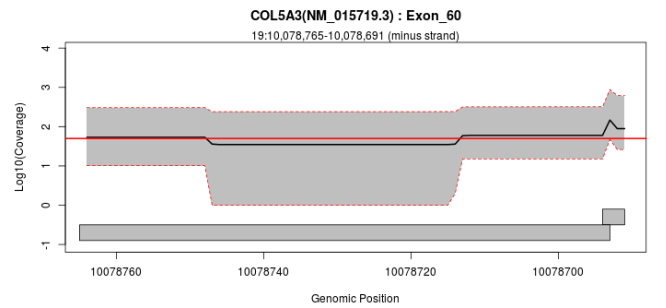
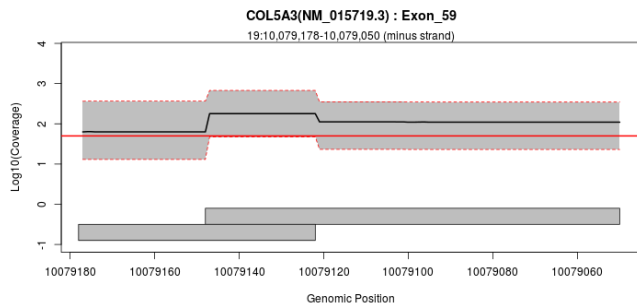
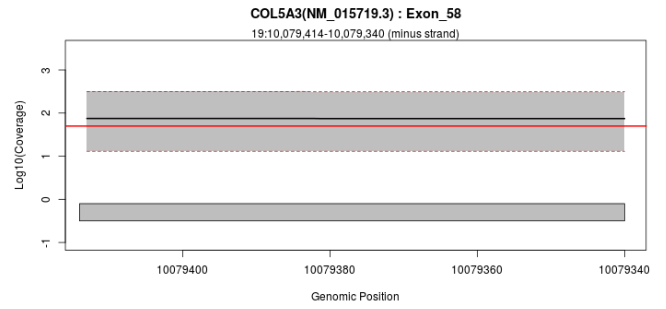
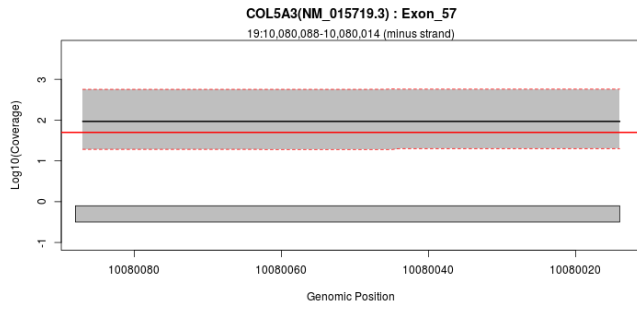
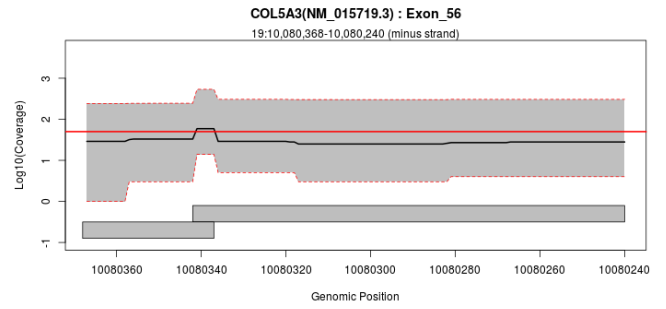
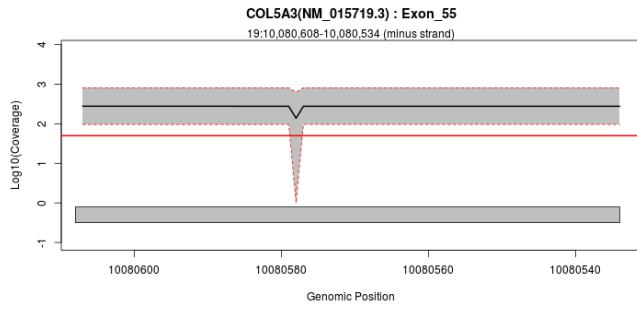


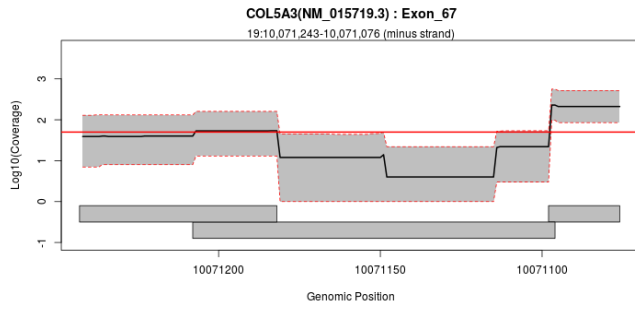
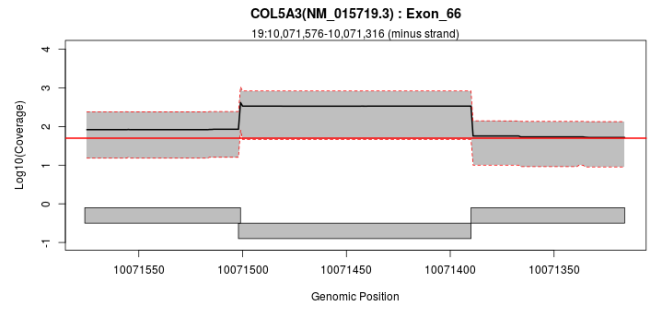
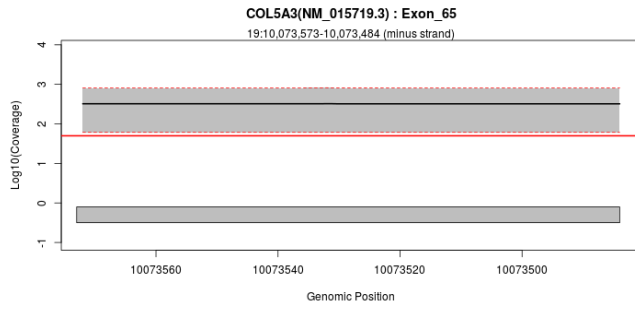




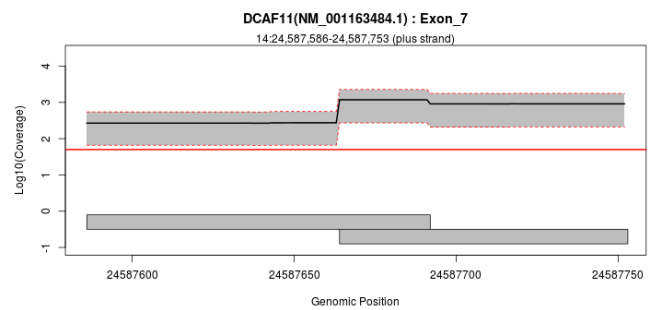
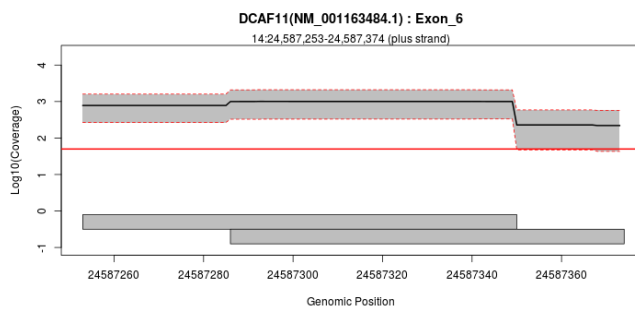
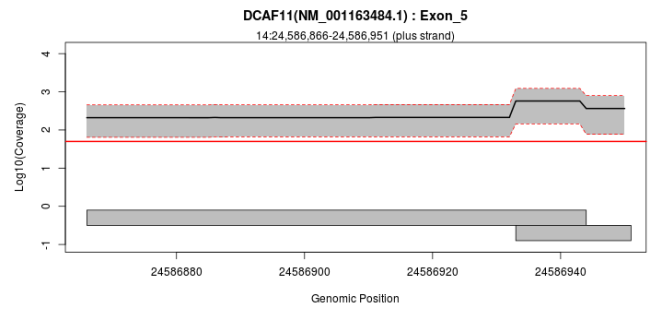
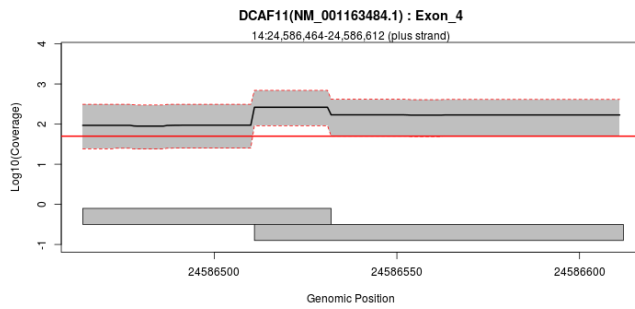
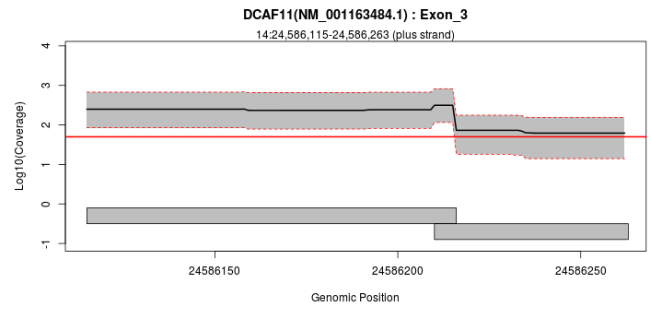
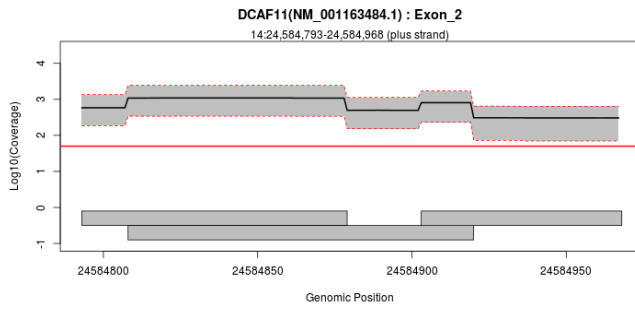


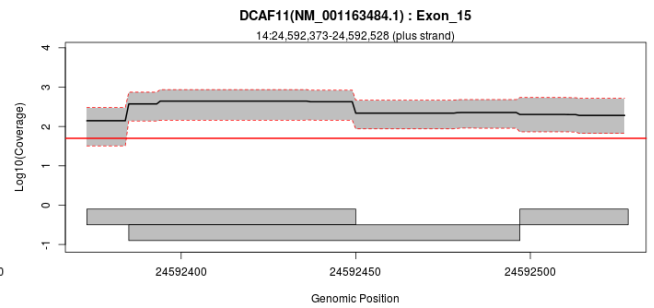
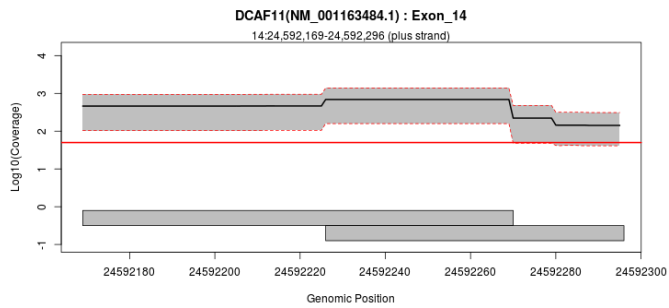
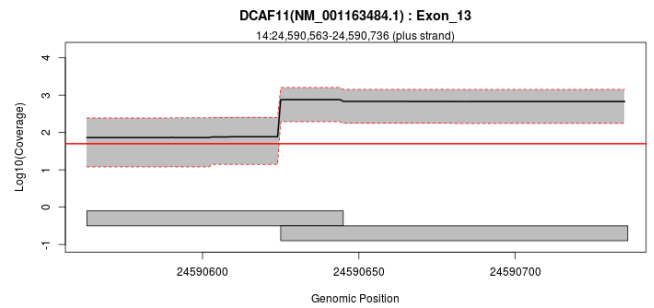
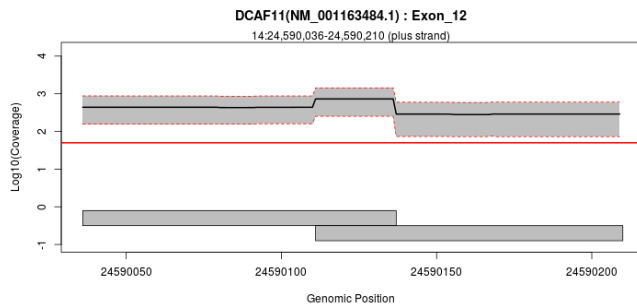
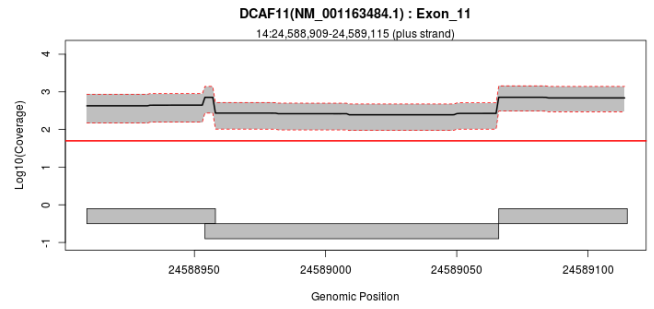
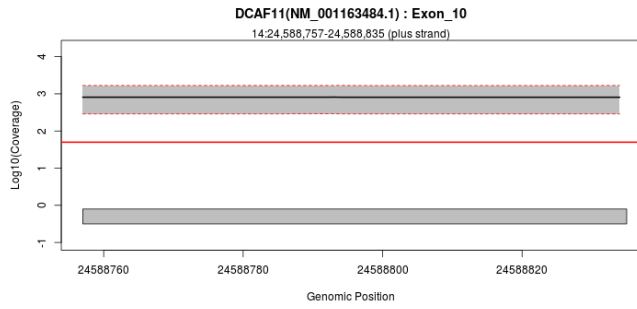
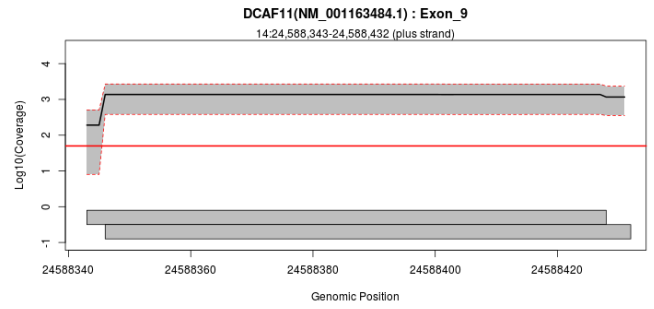
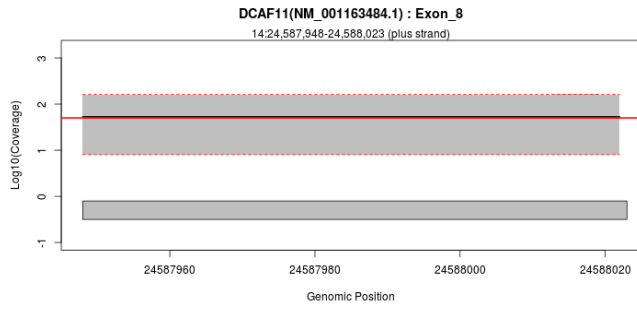




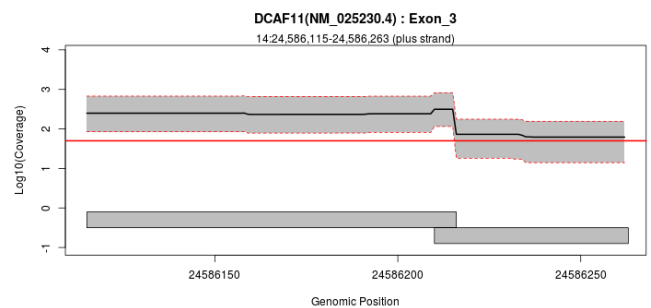
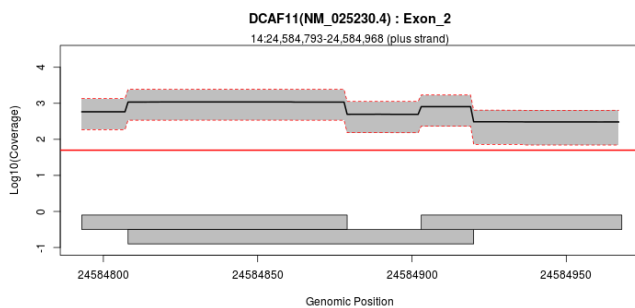


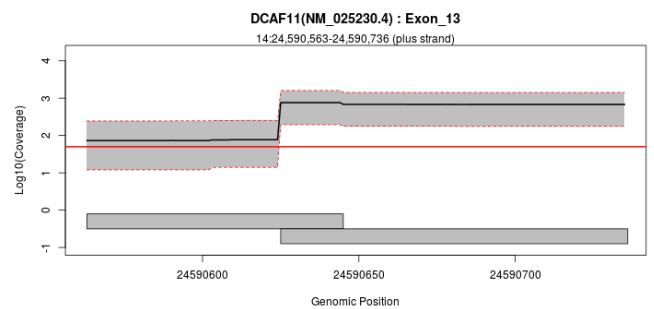
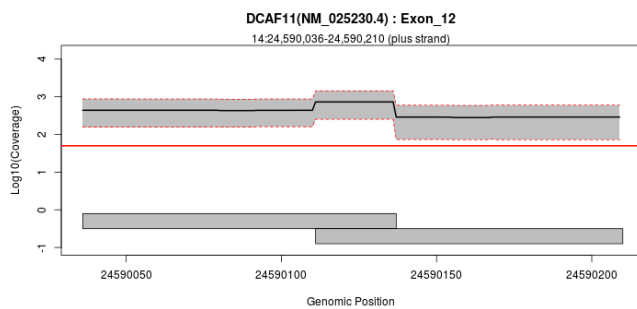
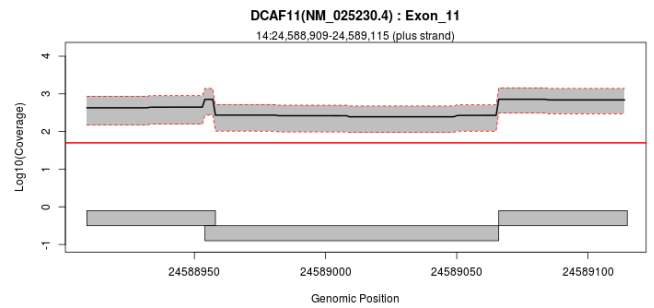
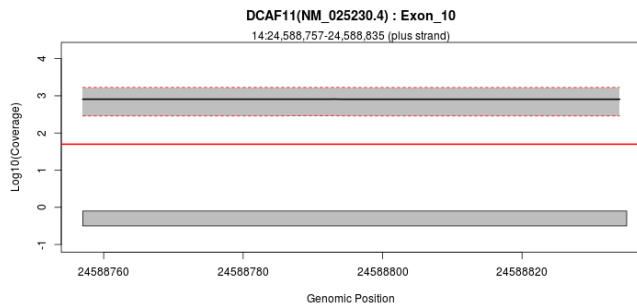
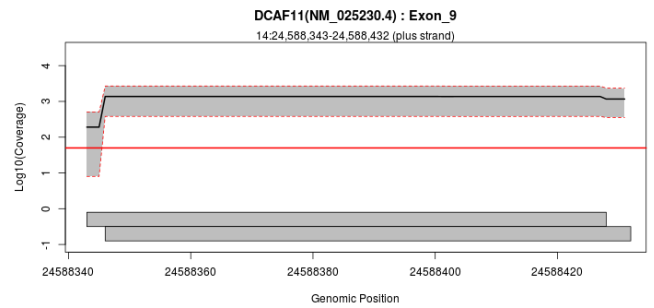
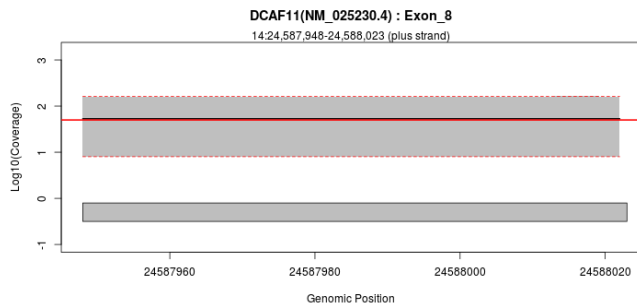
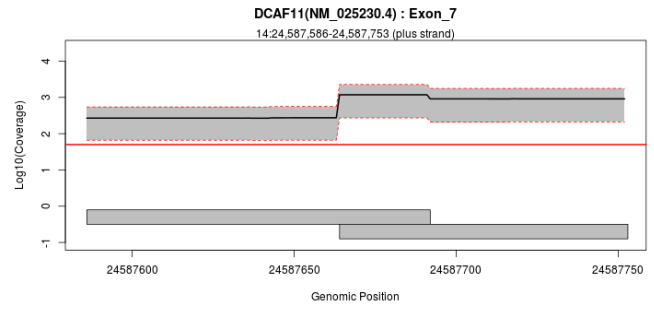
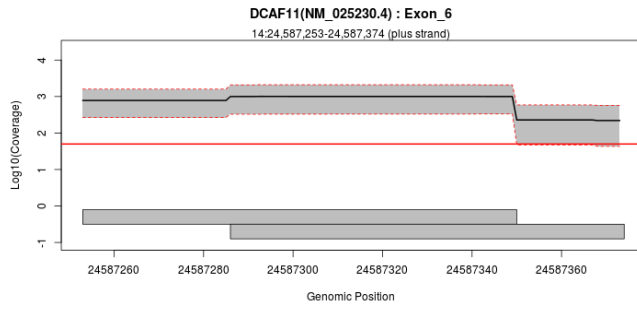
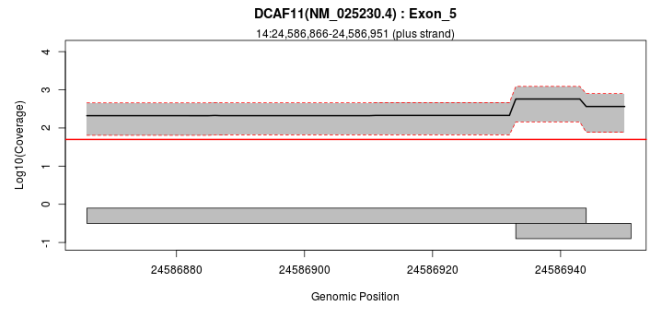
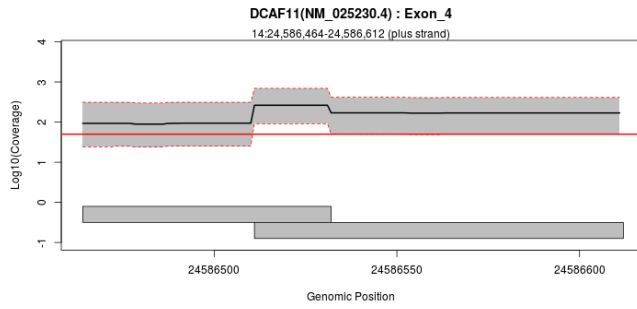
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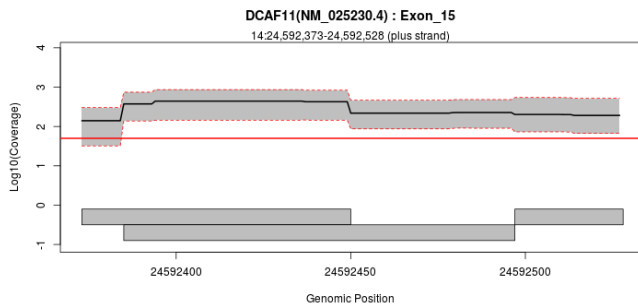
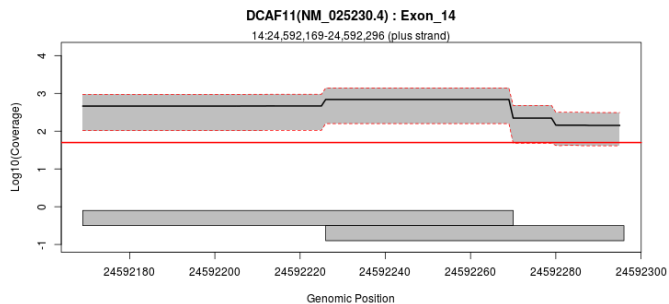




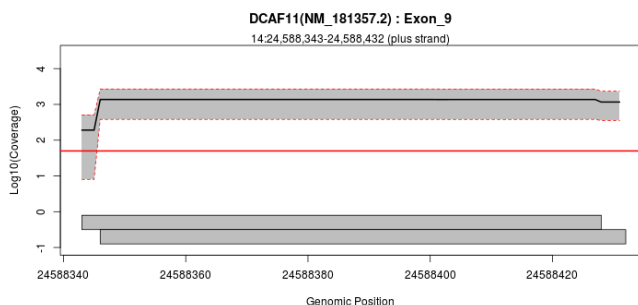
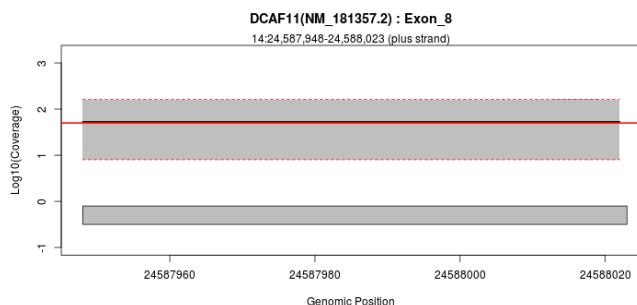
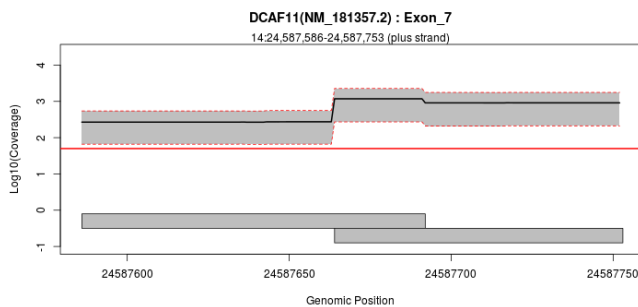
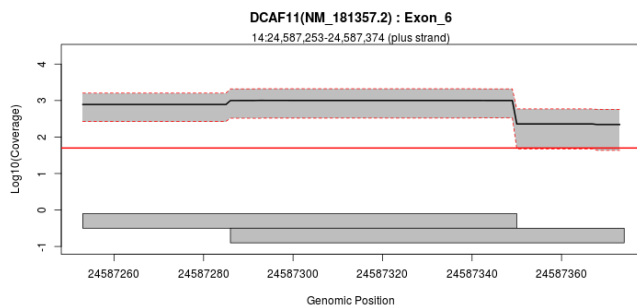
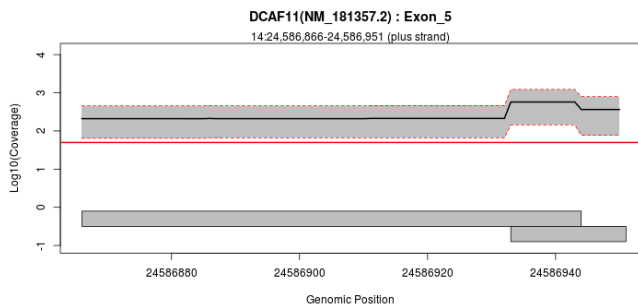
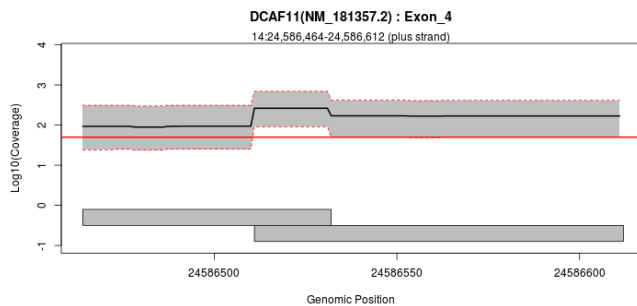
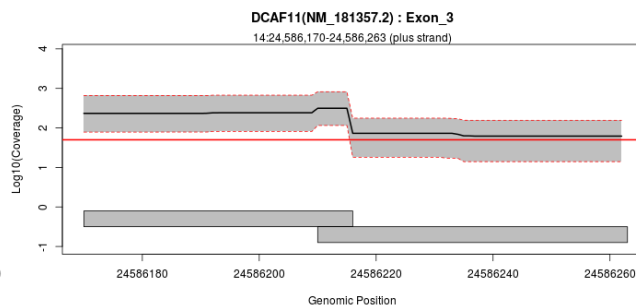
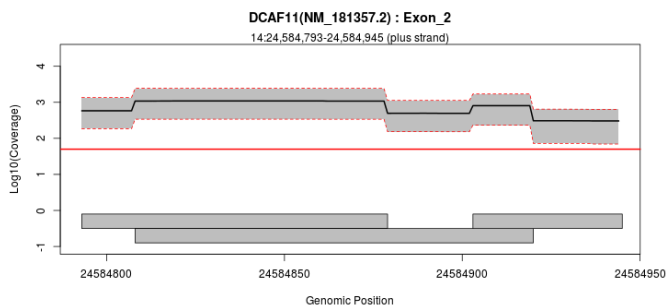
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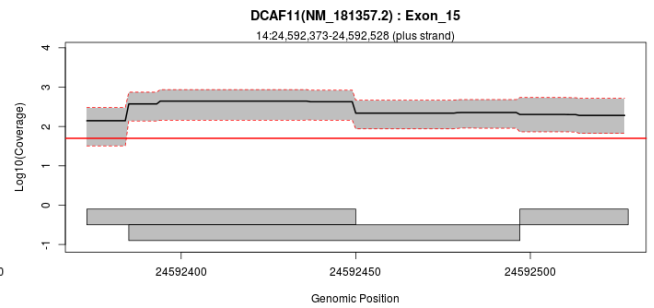
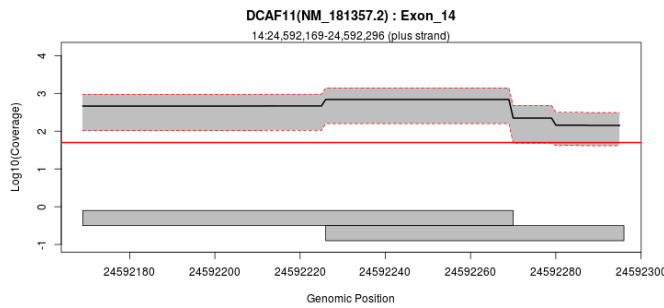
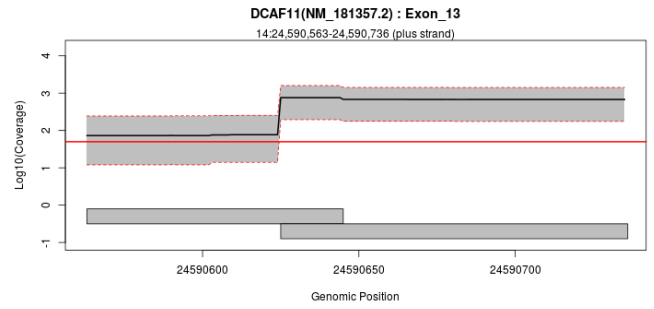
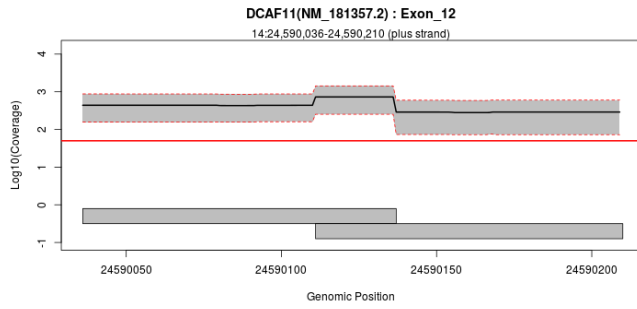
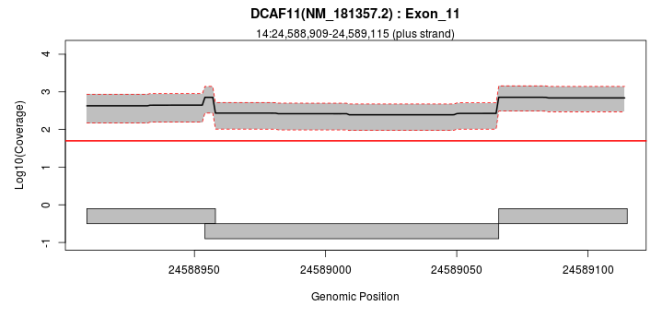
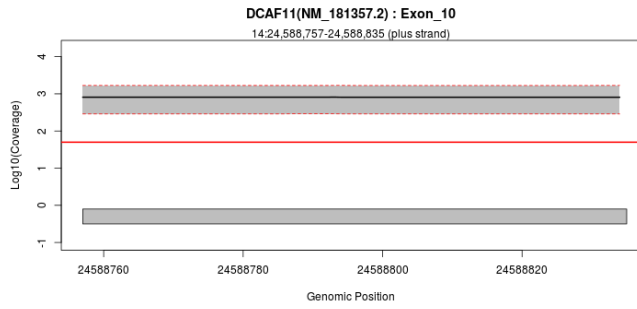




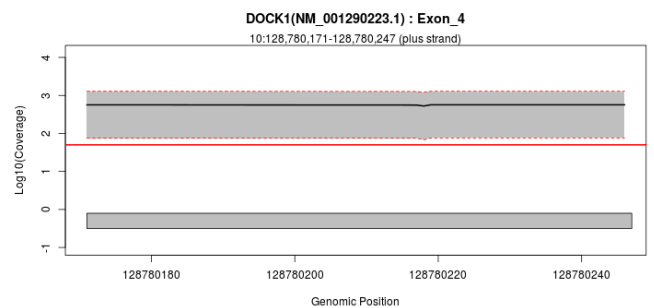
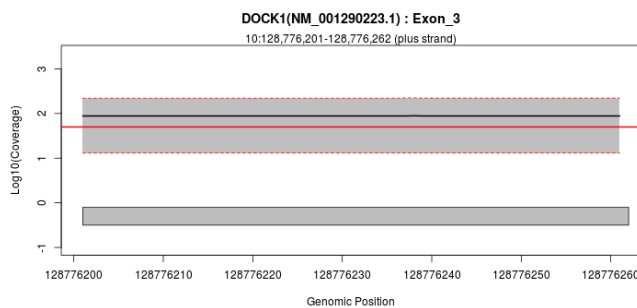
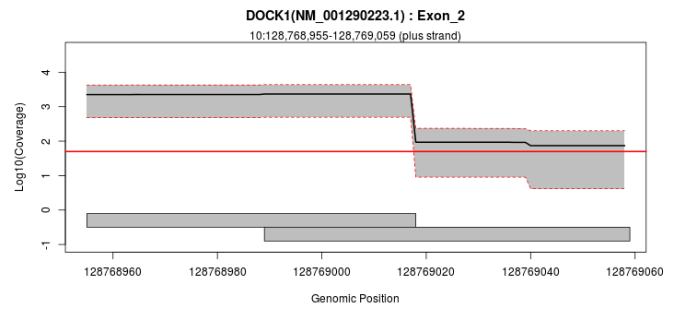
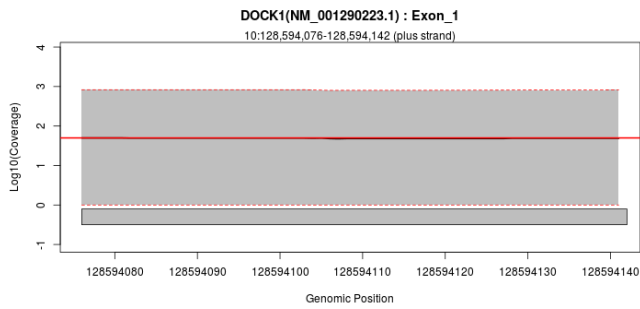


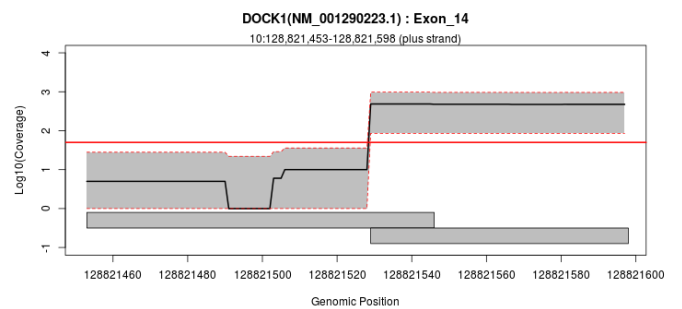
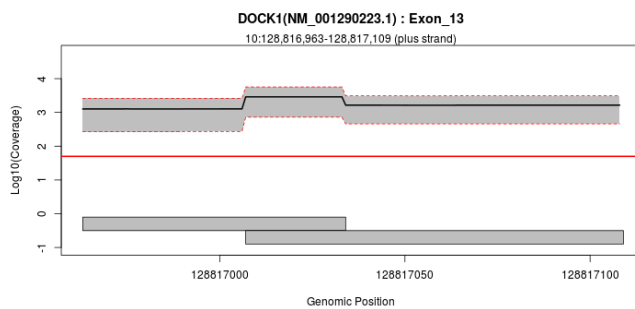
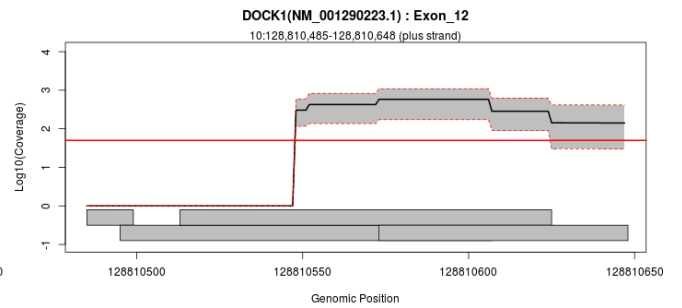
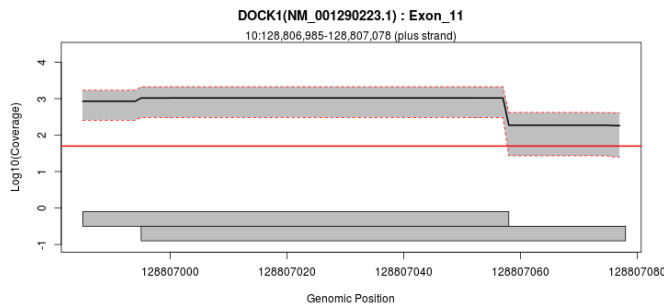
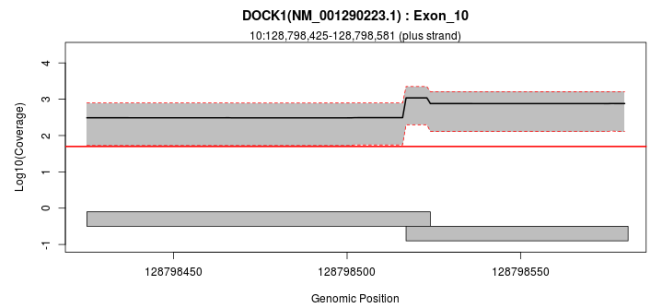
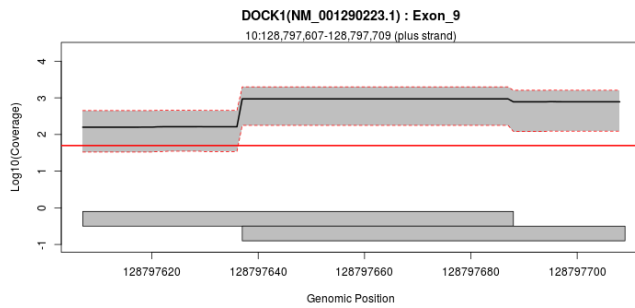
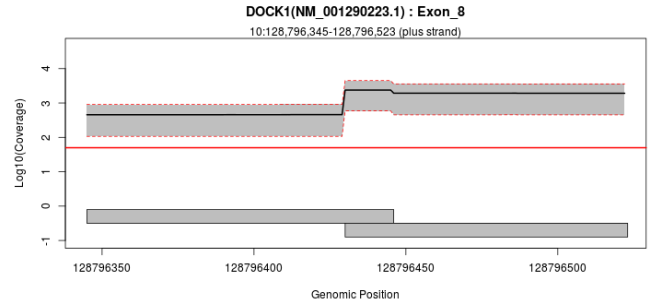
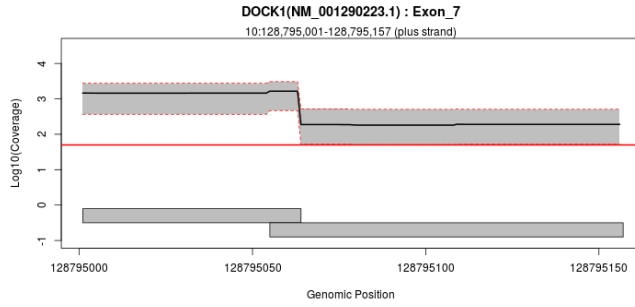
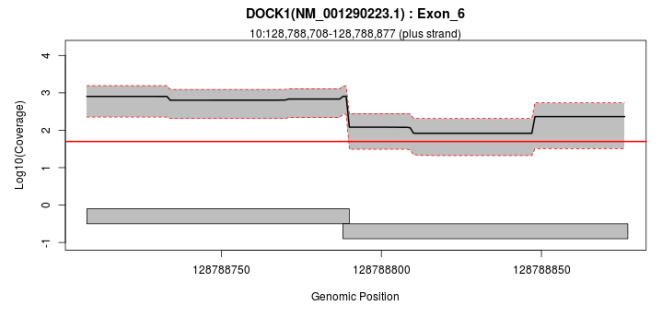
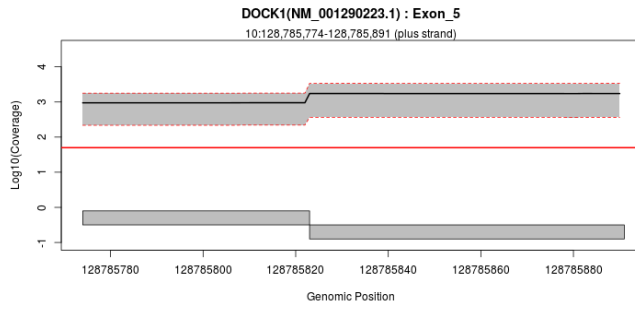
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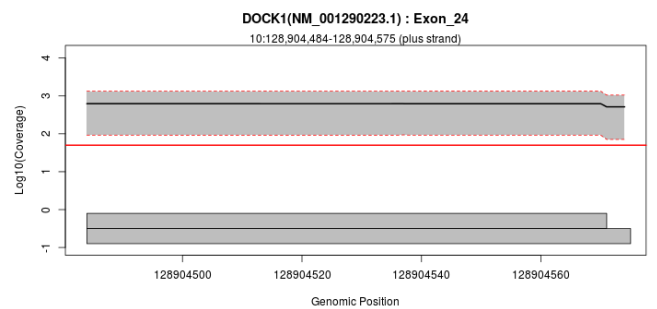
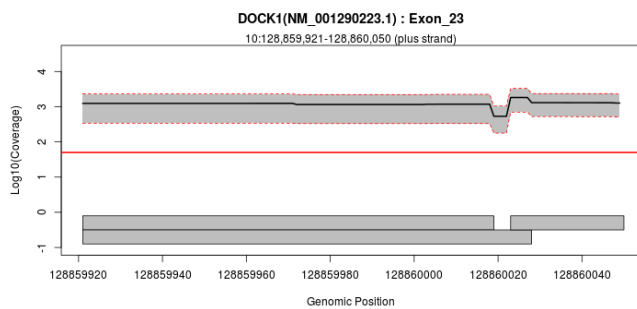
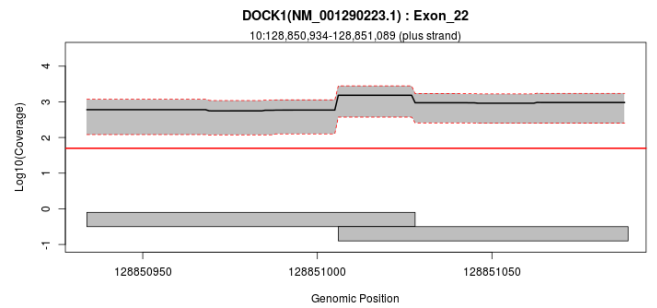
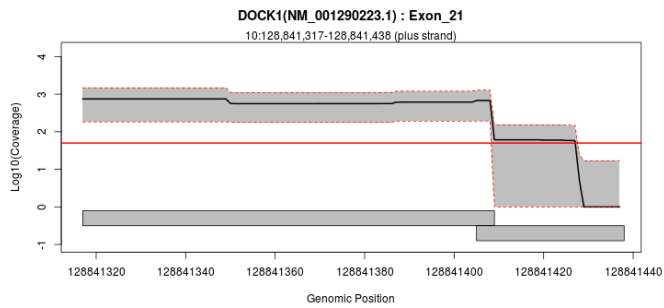
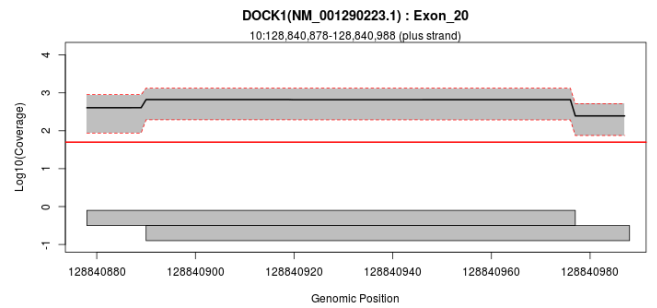
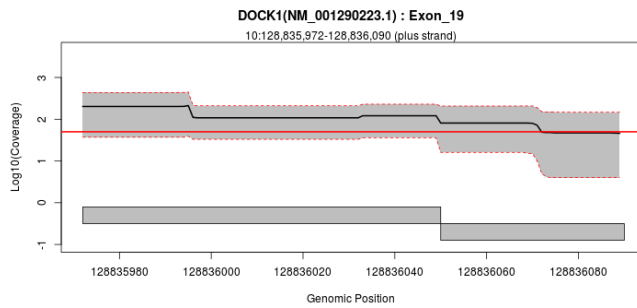
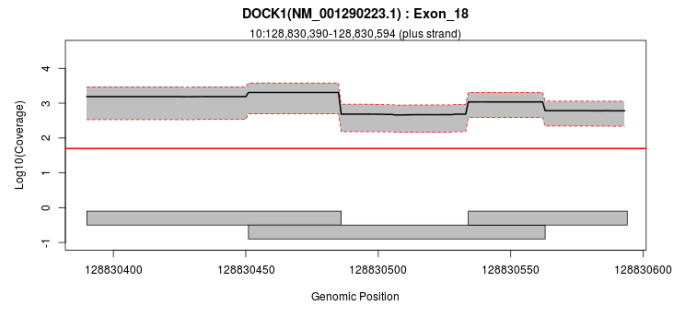
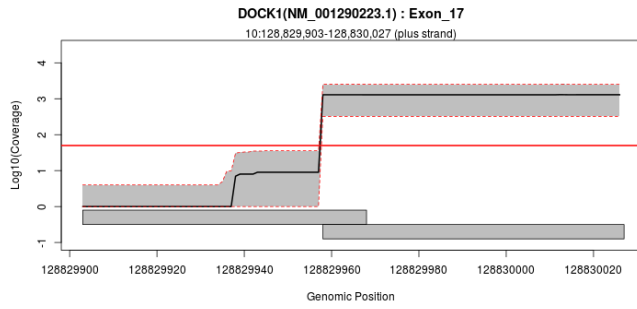
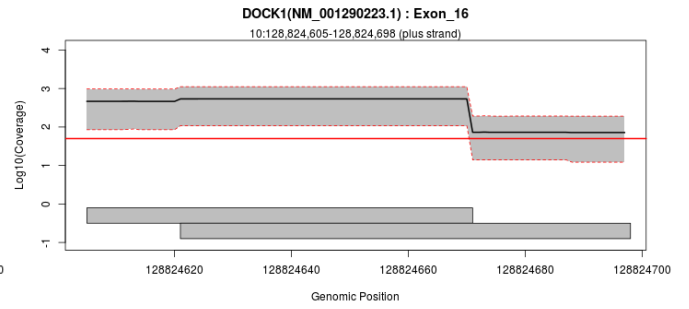
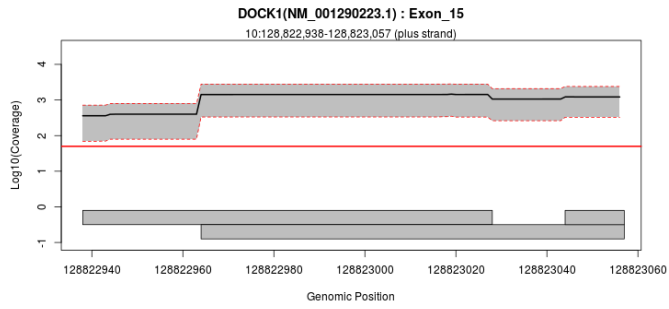


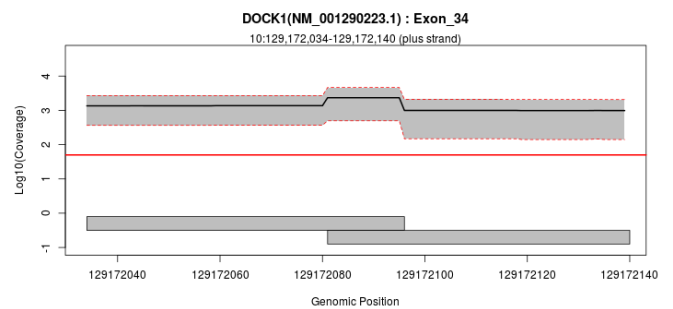
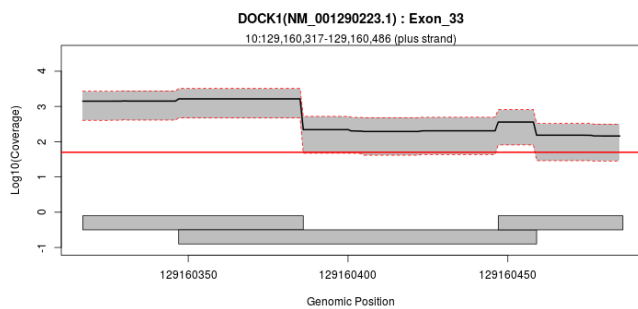
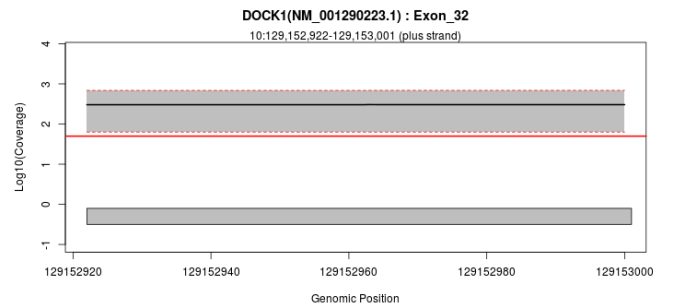
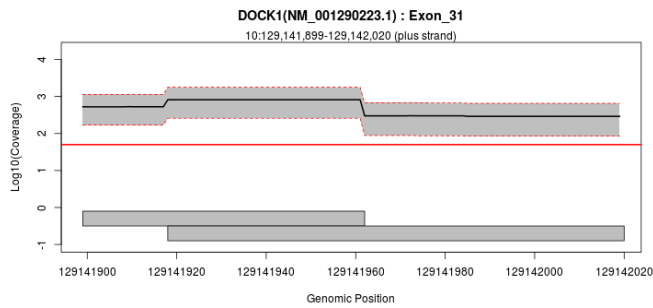
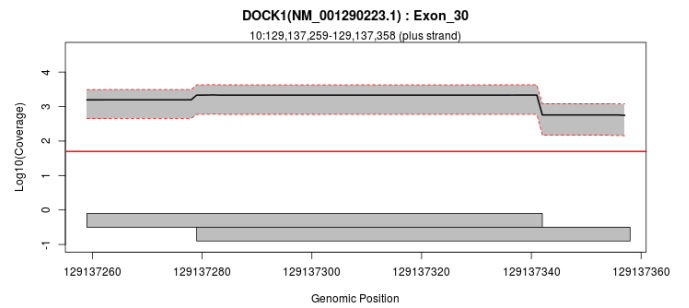
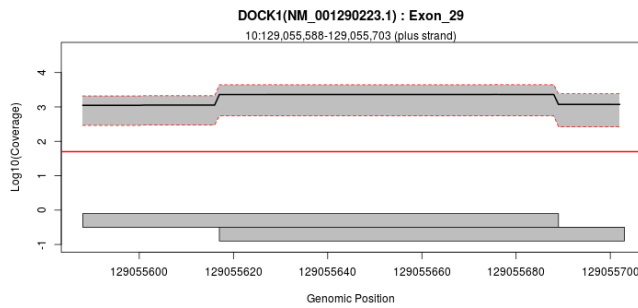
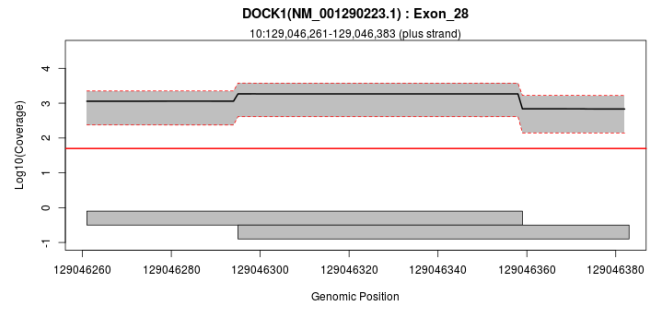
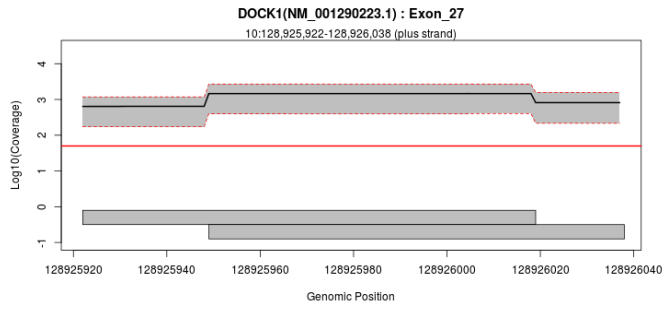
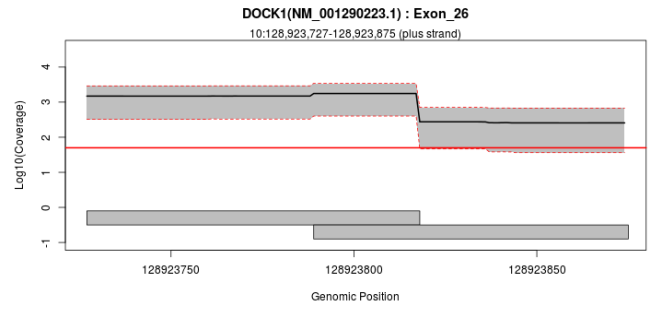
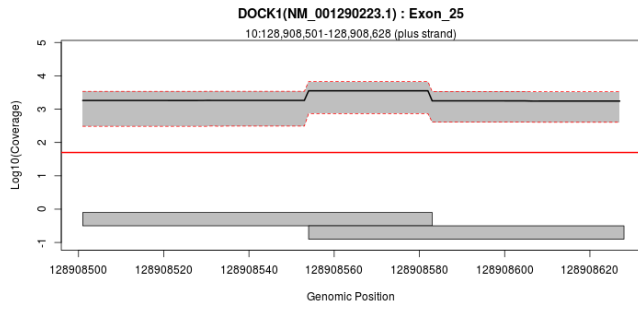
## Exon Overview: DOCK1 : NM\_001290223.1

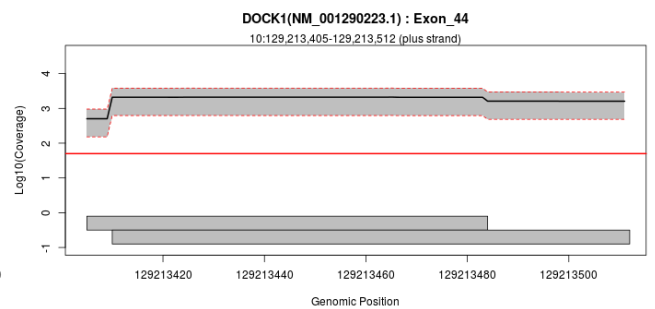
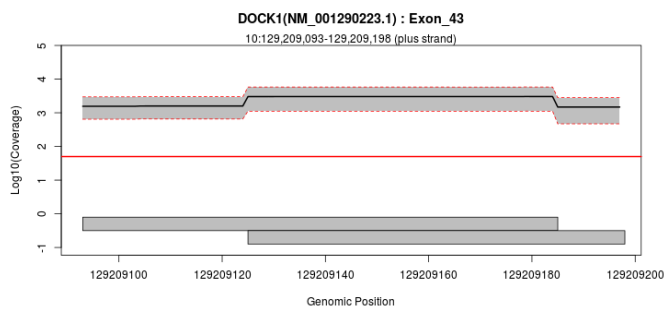
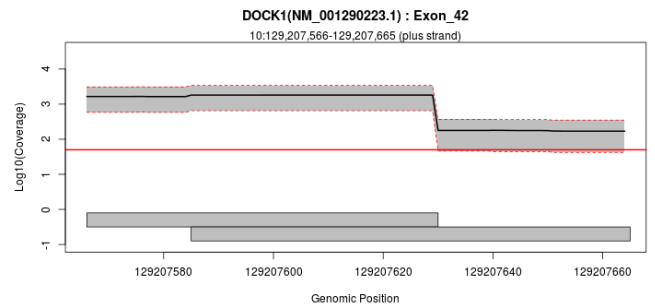
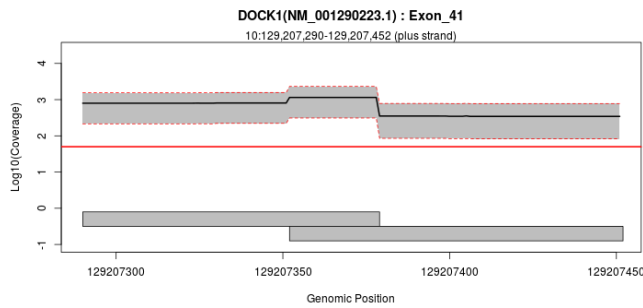
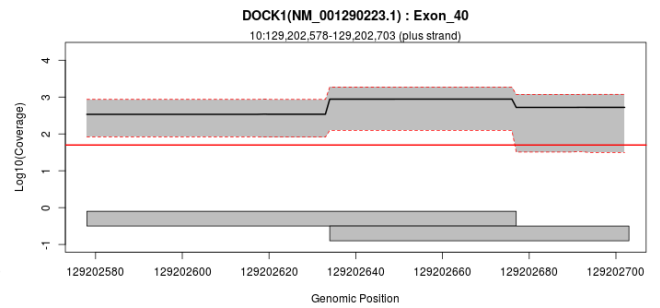
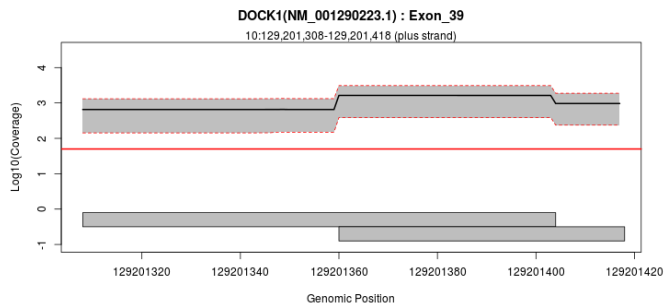
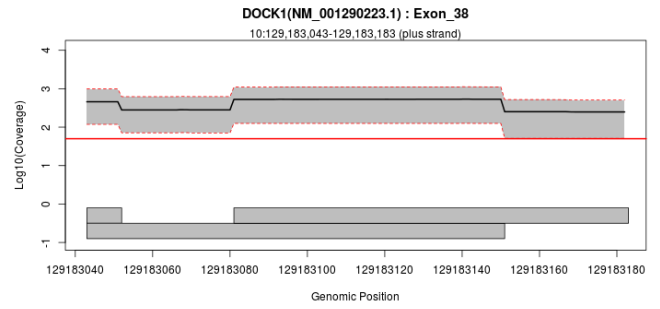
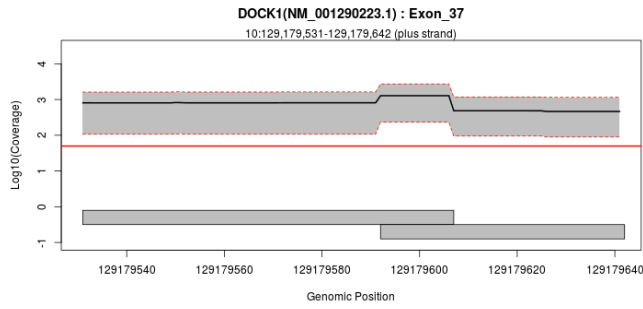
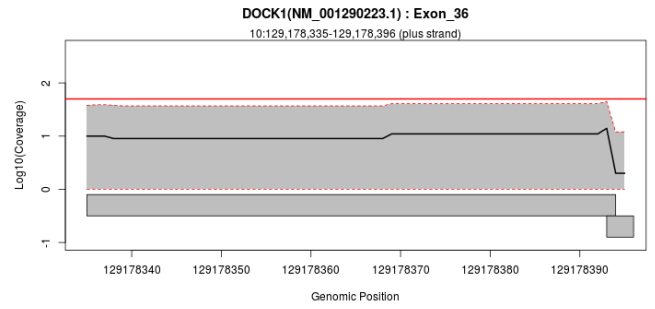
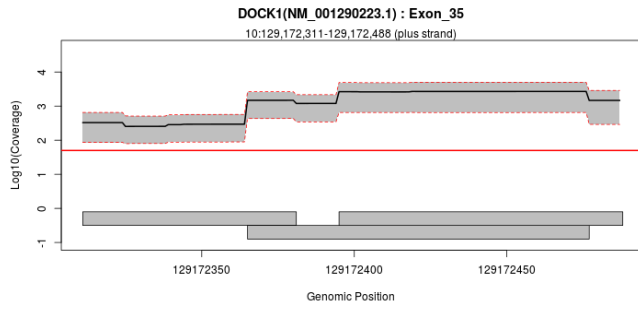


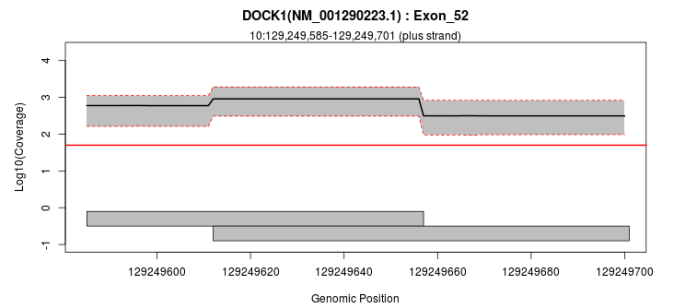
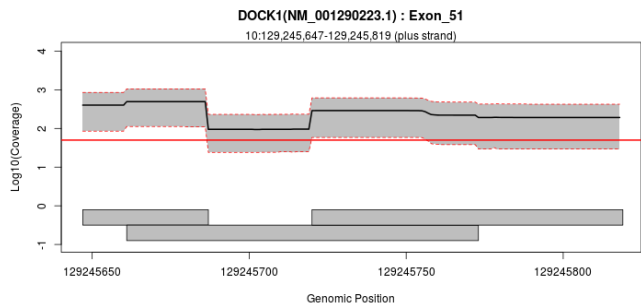
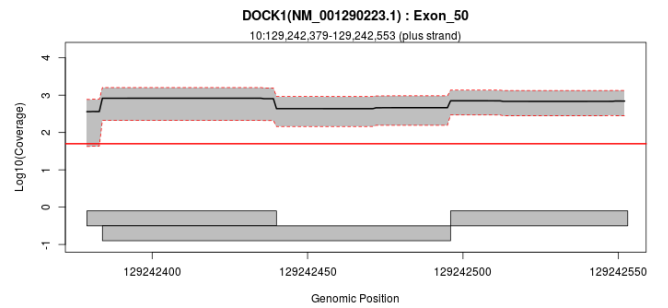
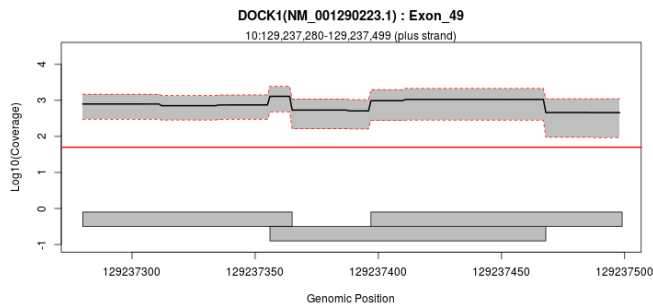
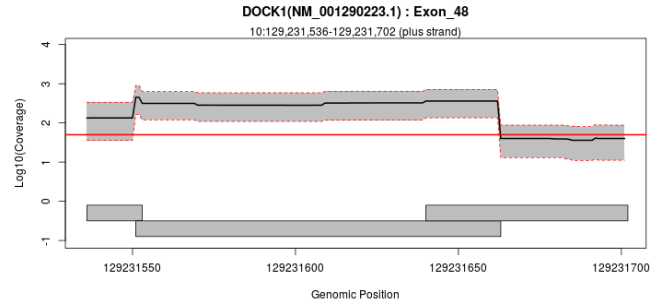
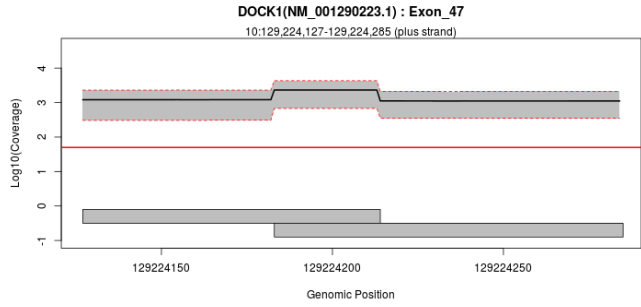
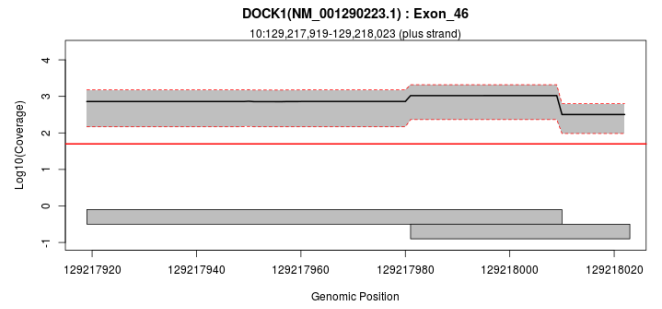
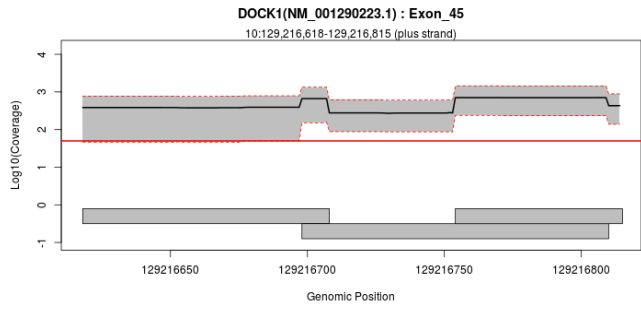




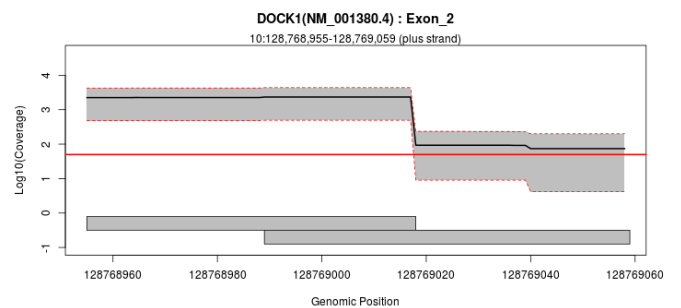
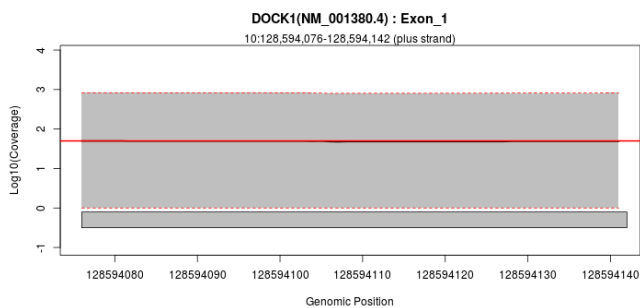


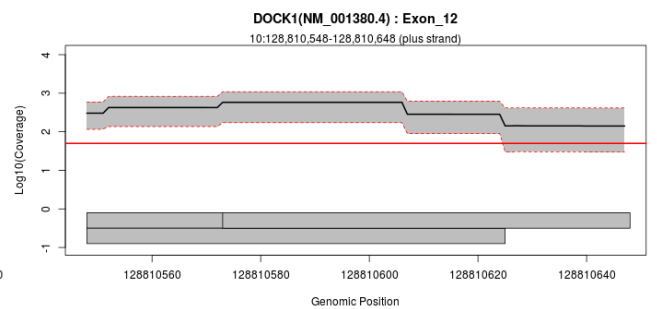
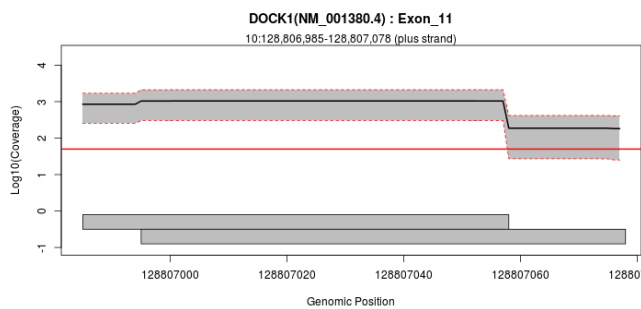
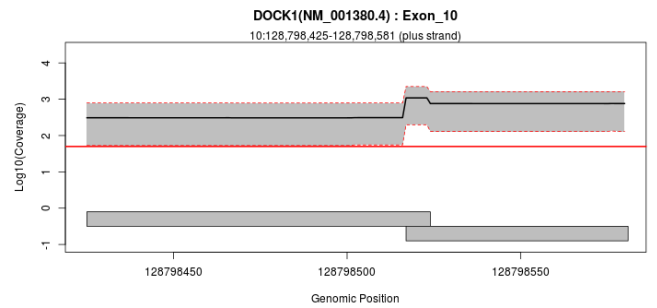
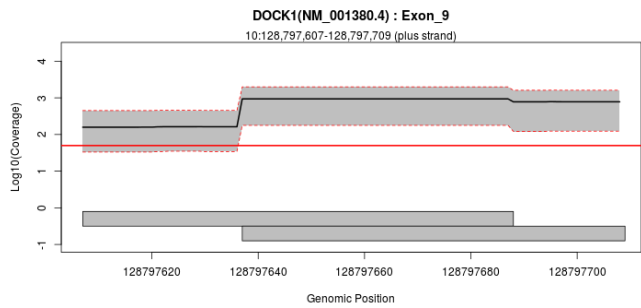
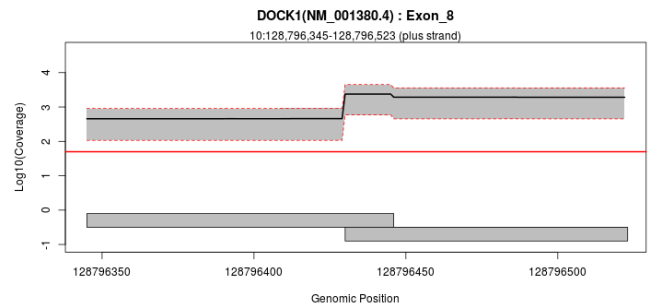
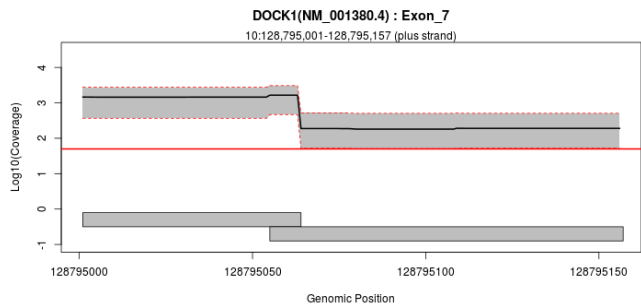
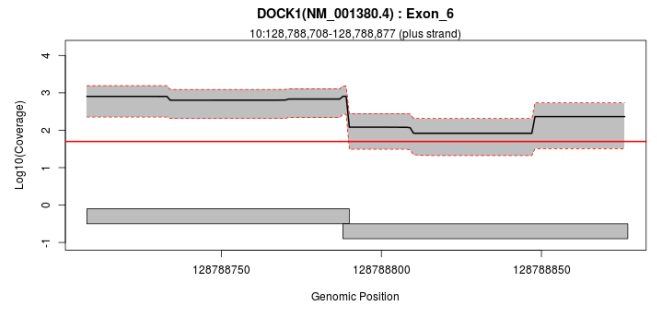
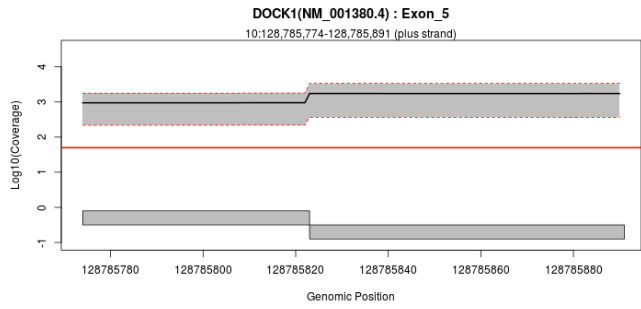
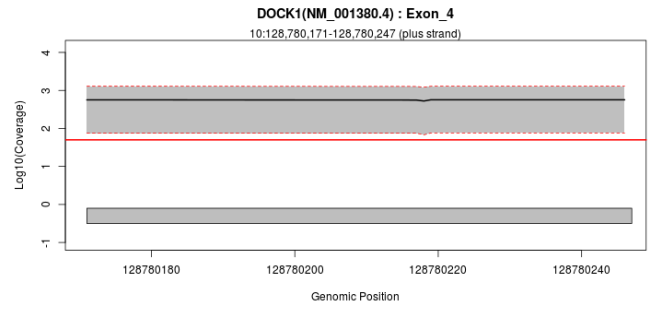
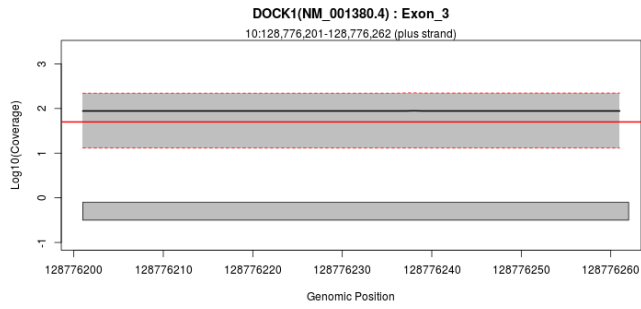


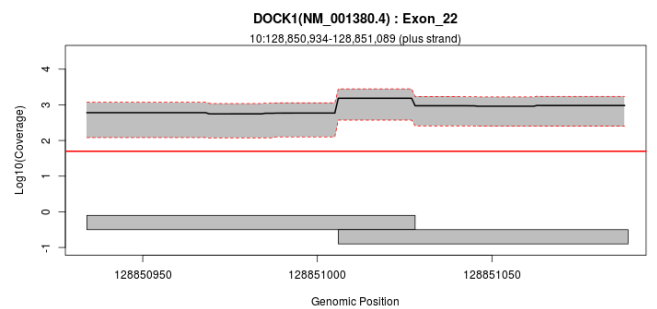
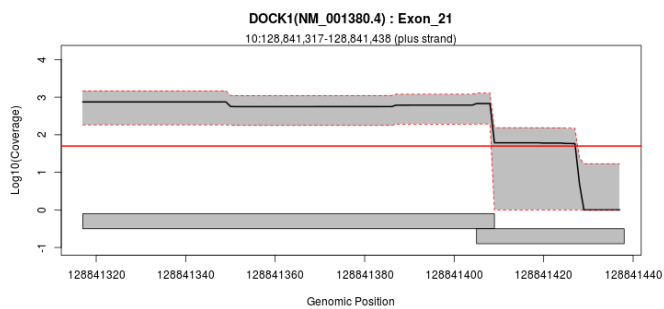
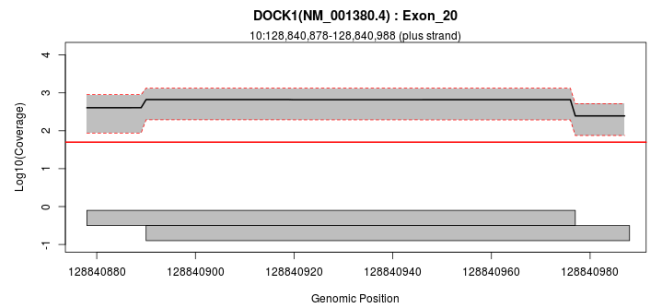
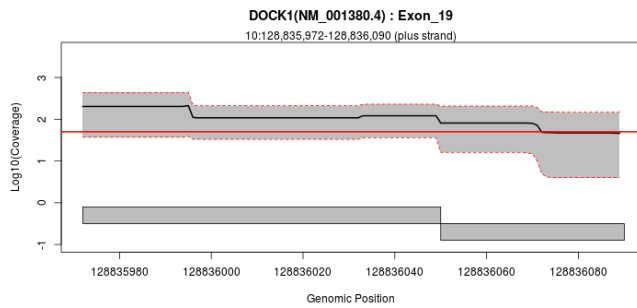
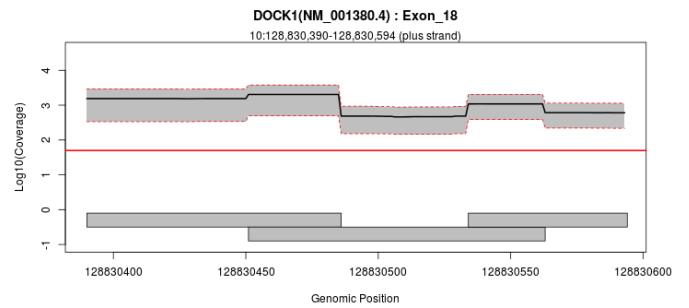
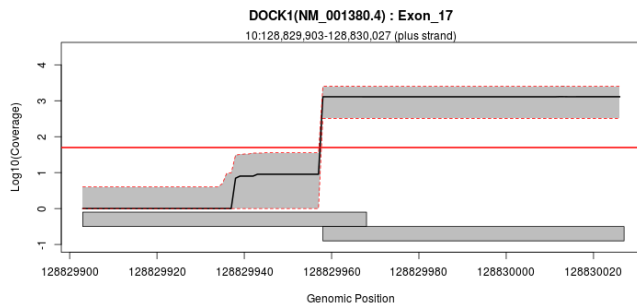
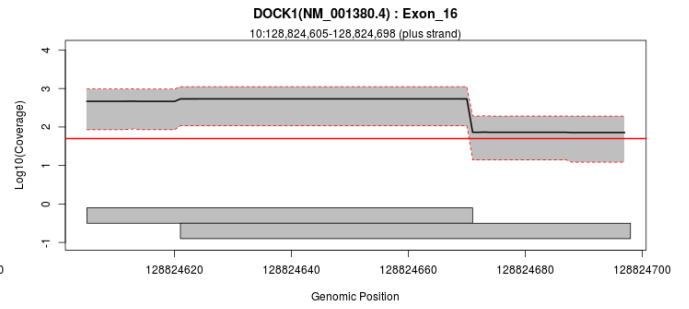
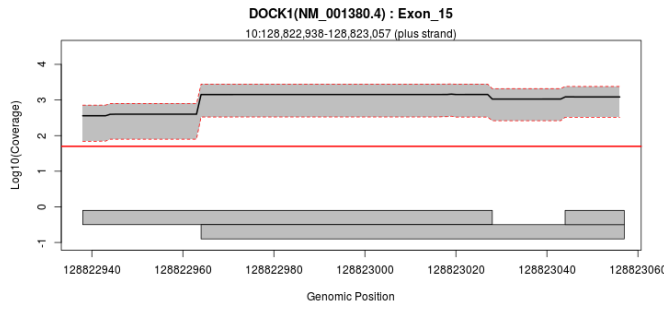
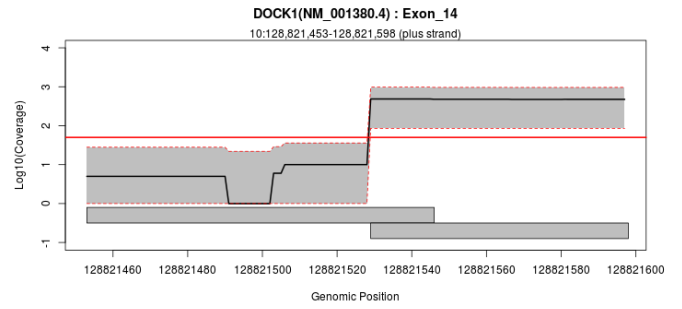
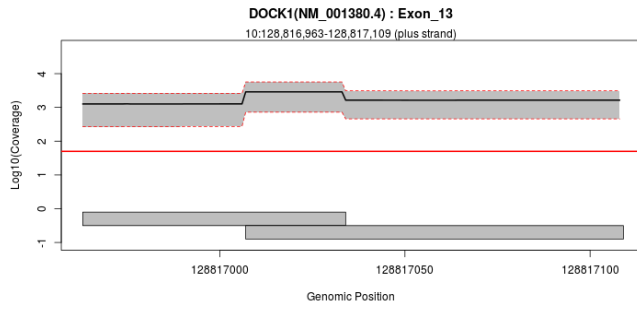


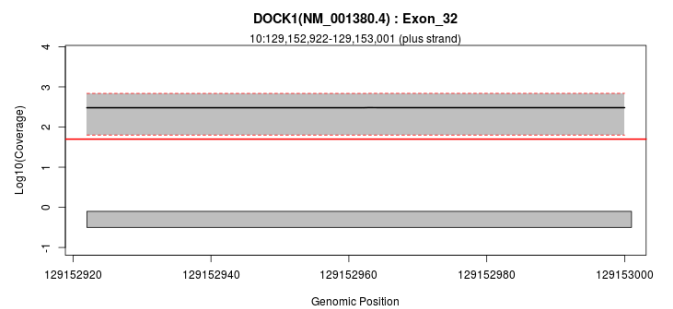
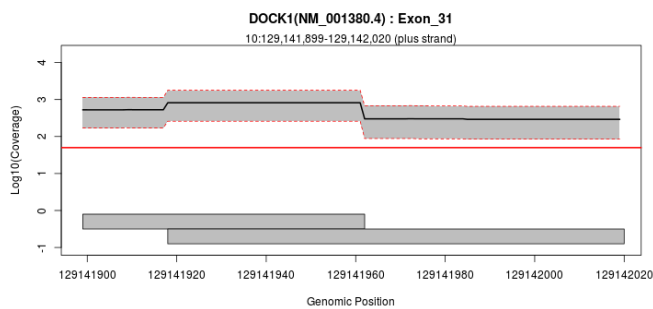
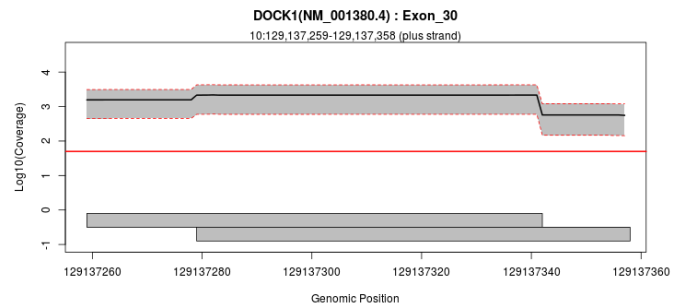
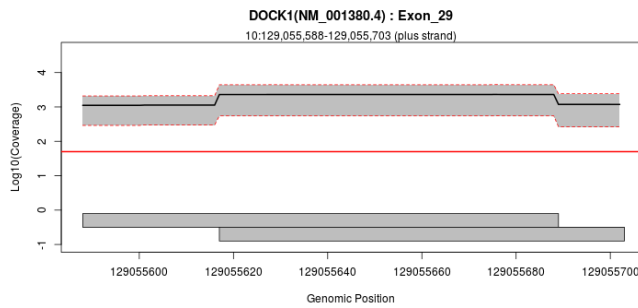
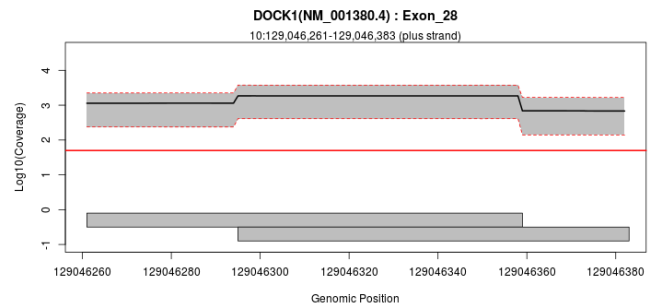
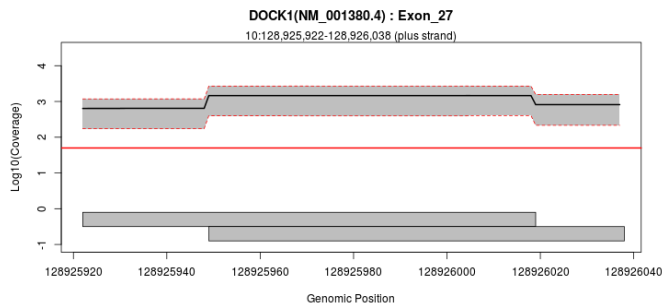
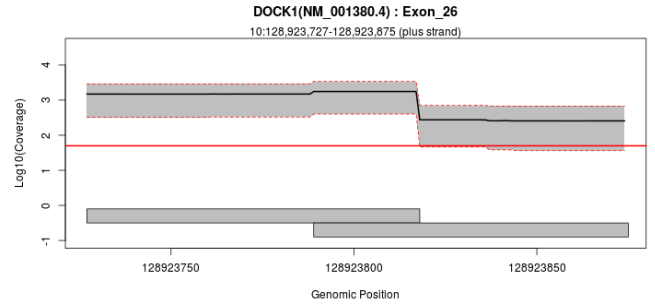
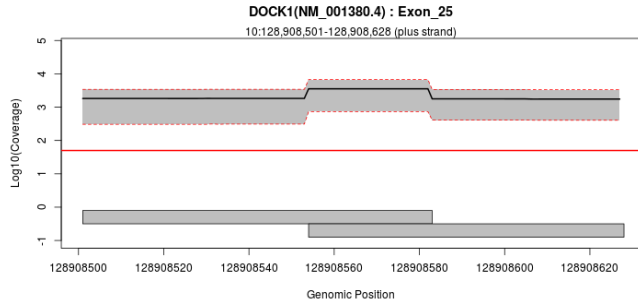
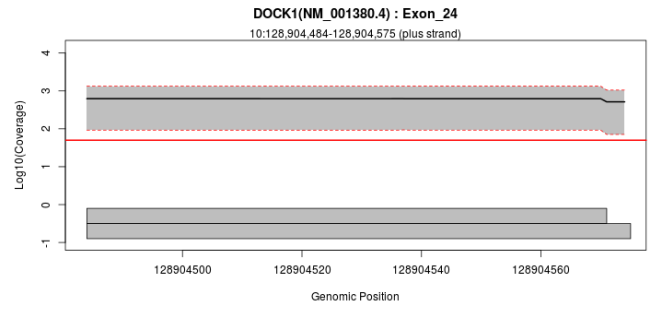
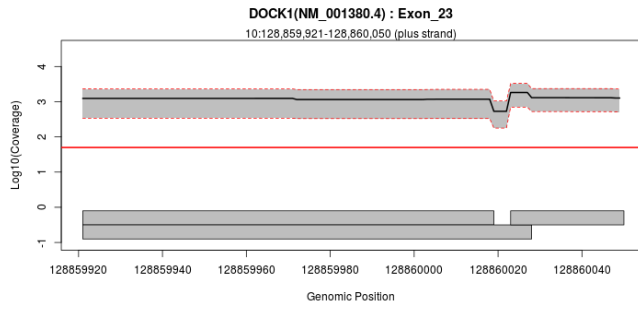


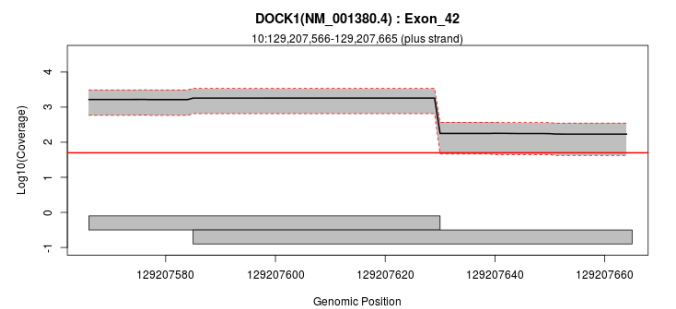
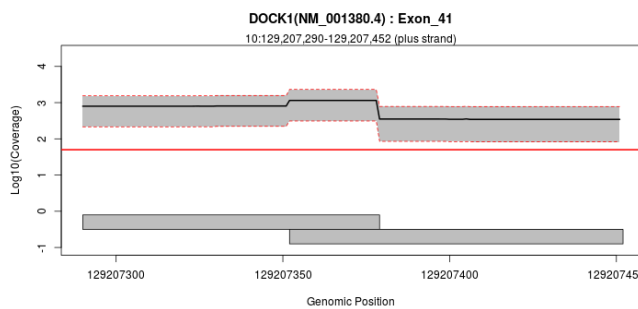
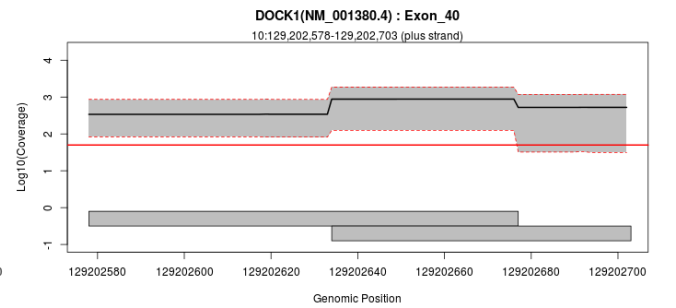
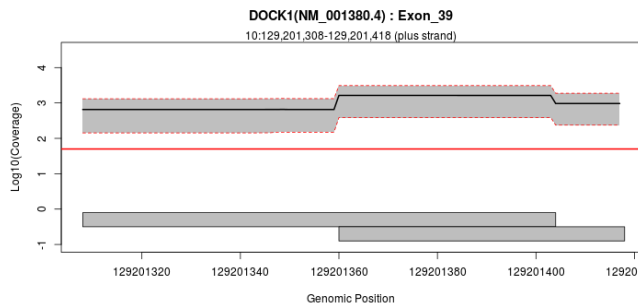
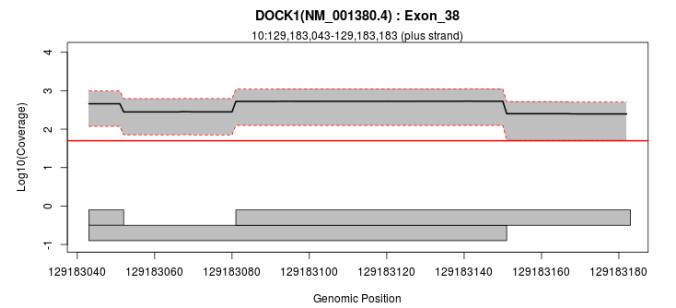
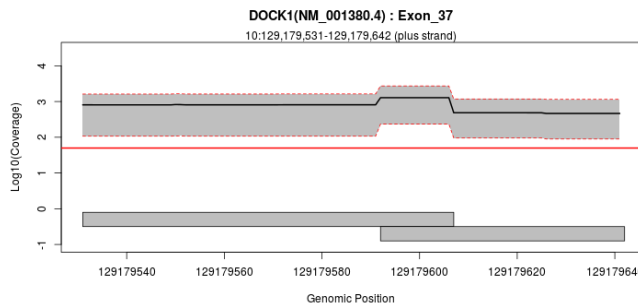
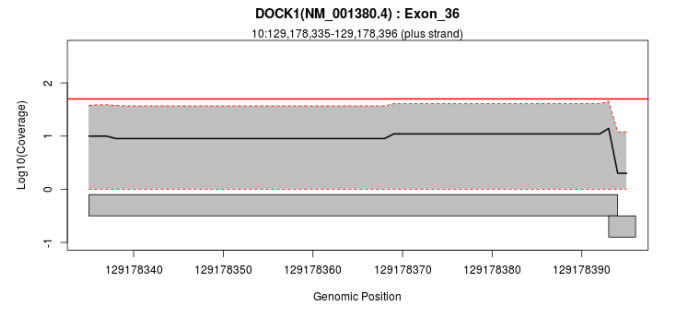
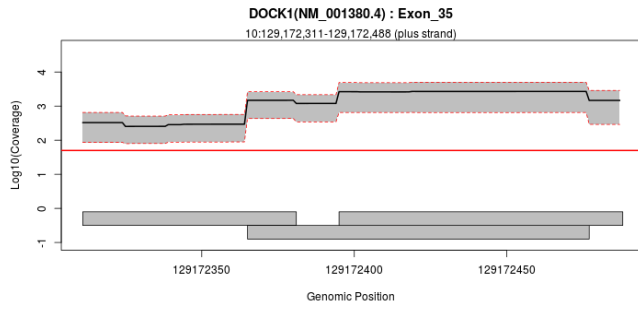
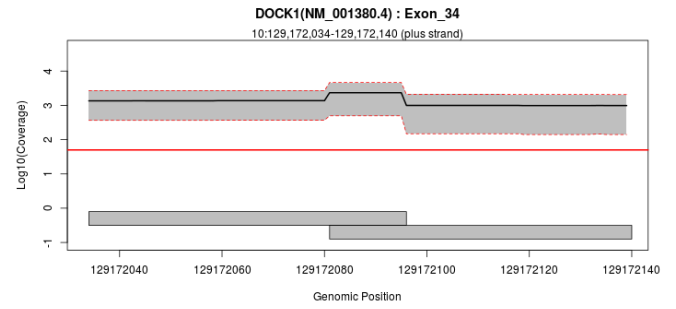
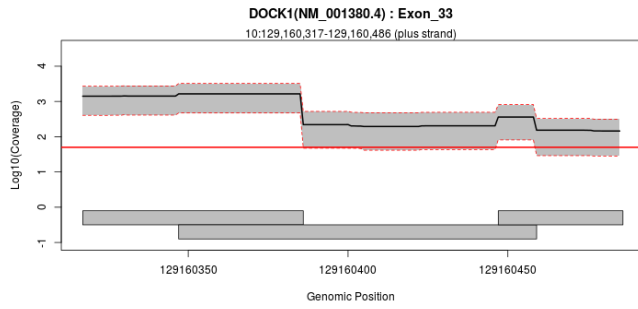
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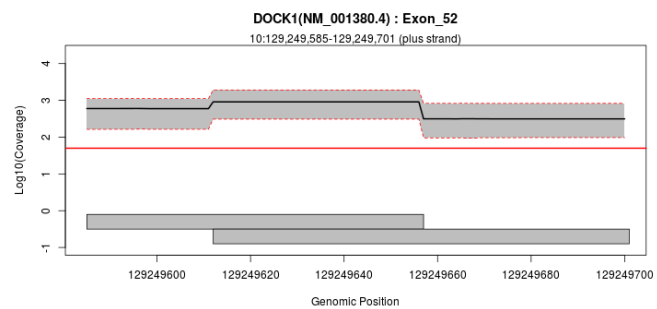
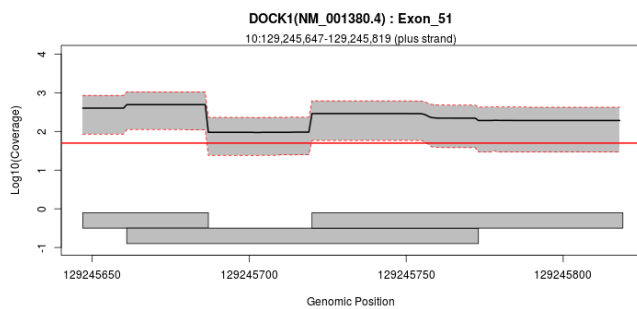
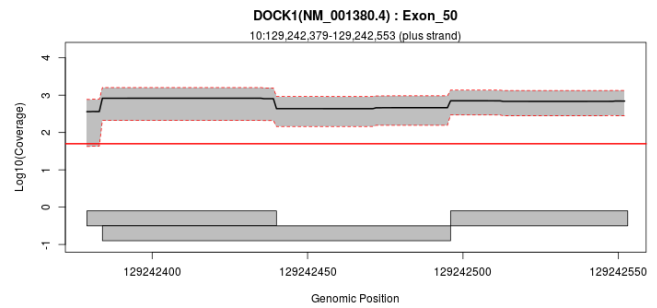
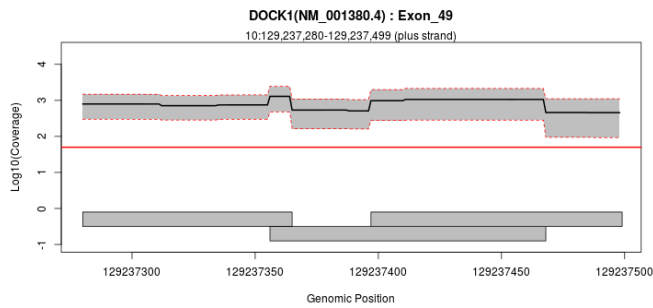
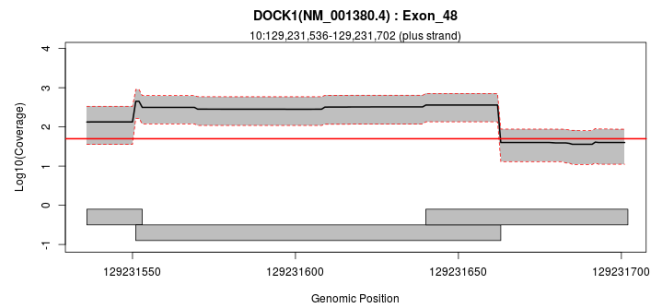
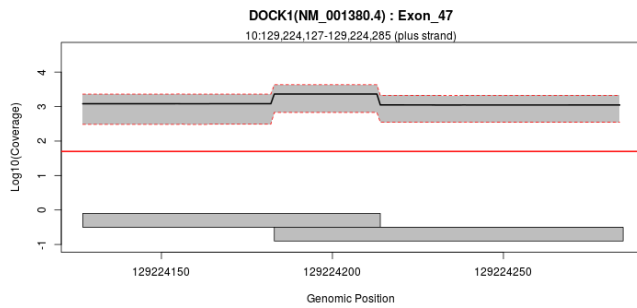
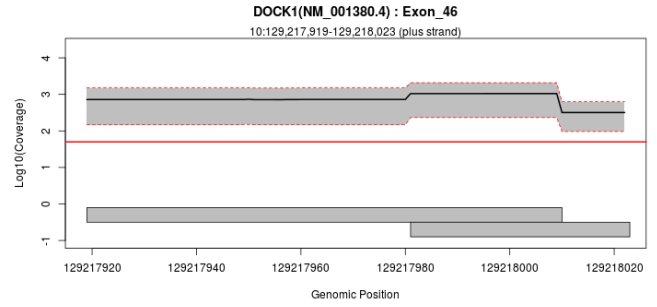
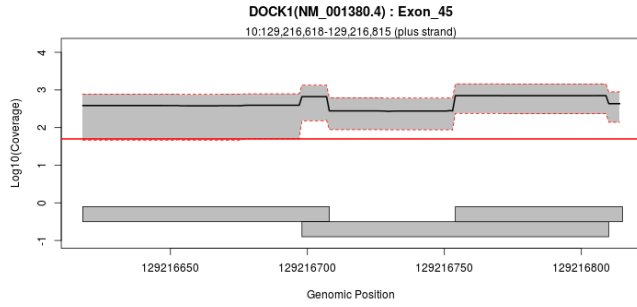
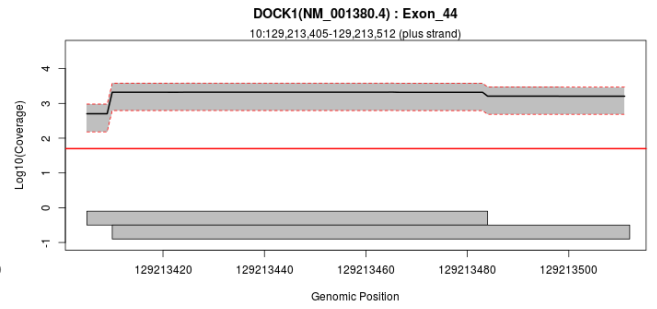
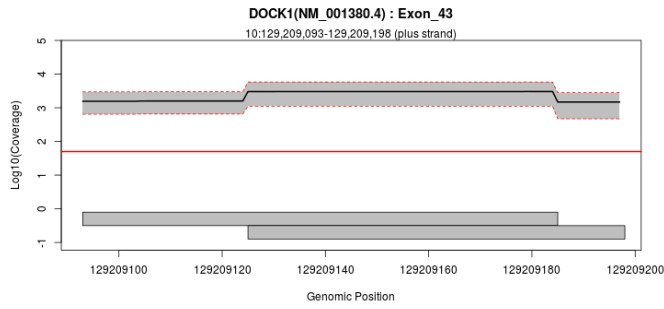




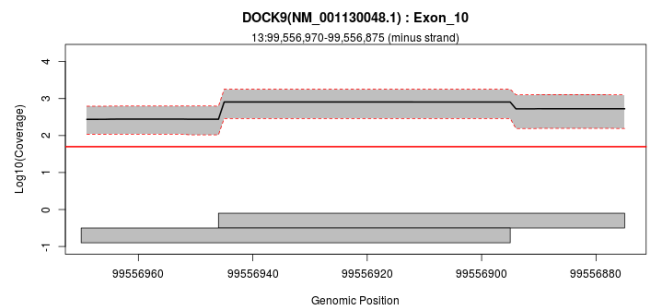
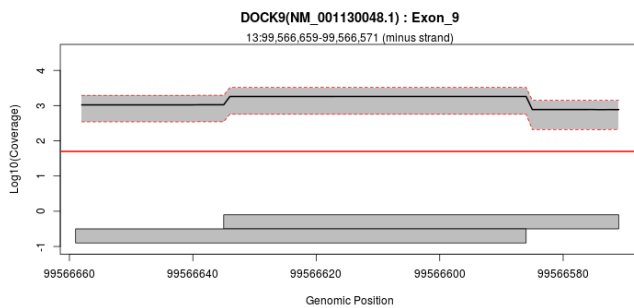
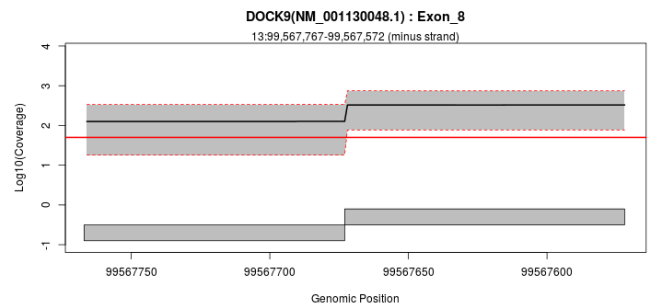
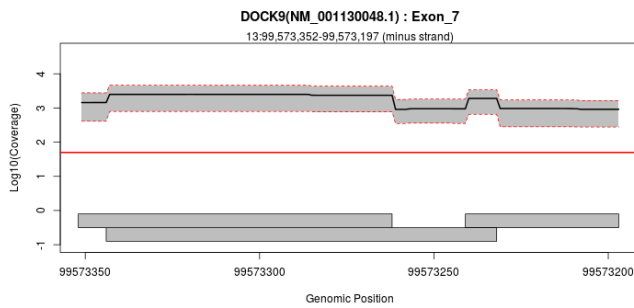
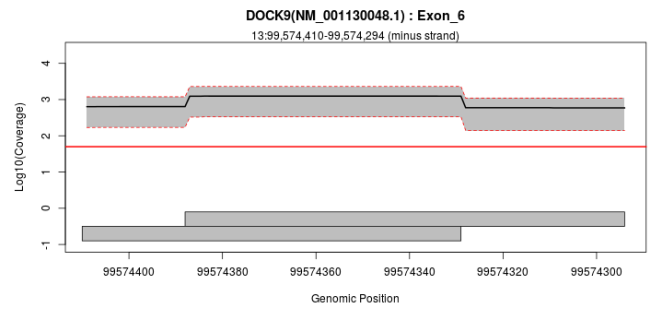
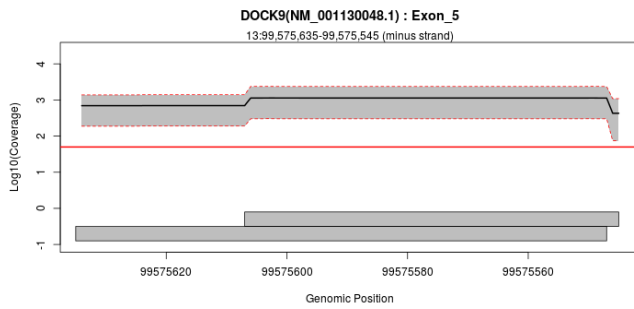
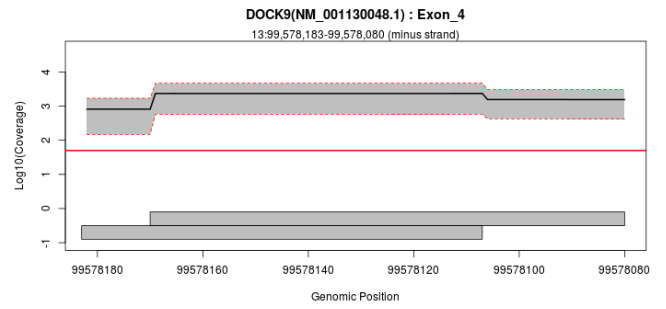
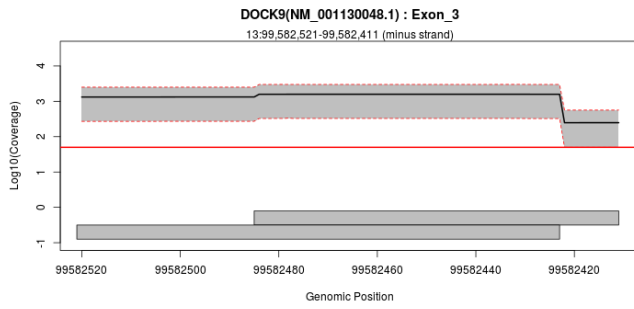
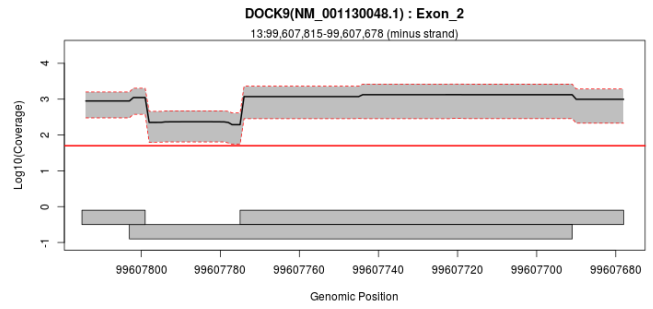
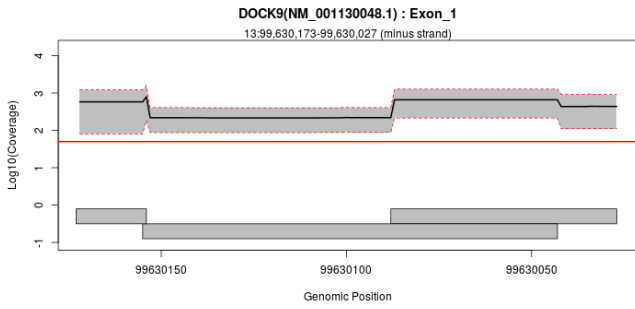


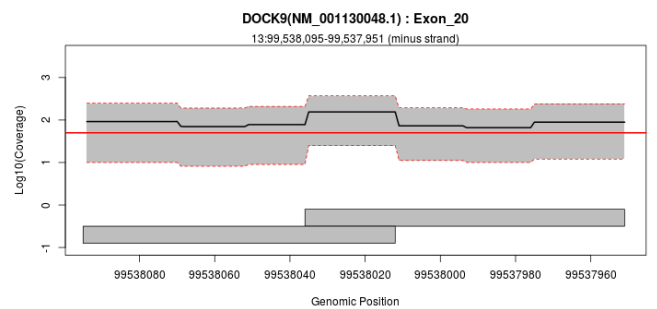
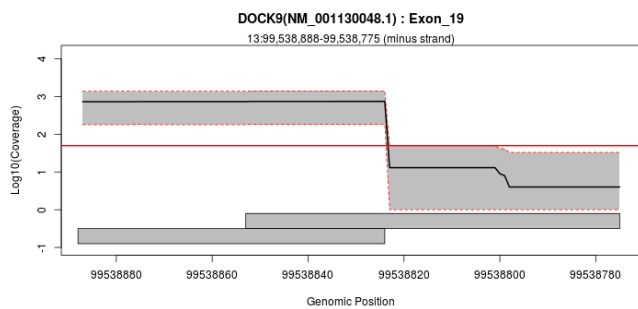
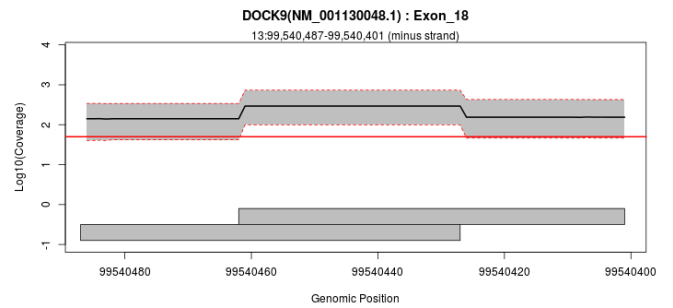
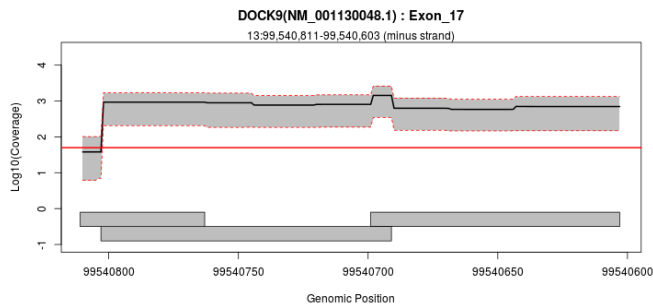
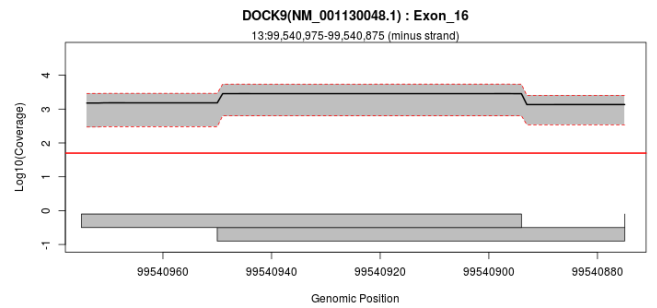
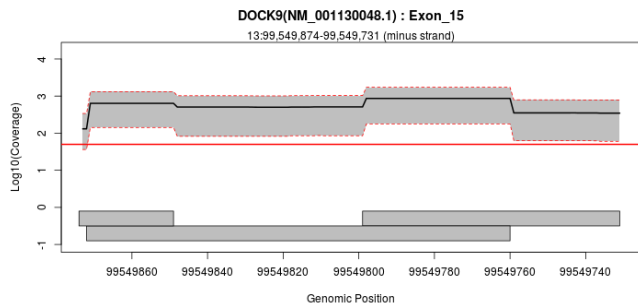
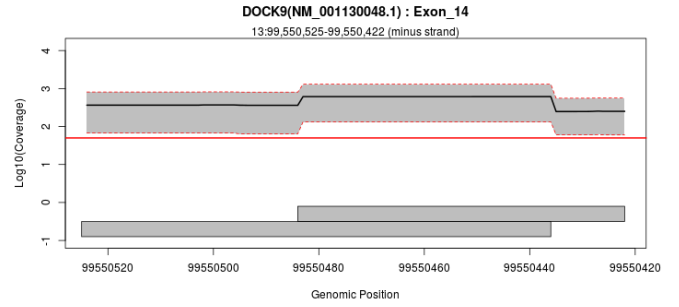
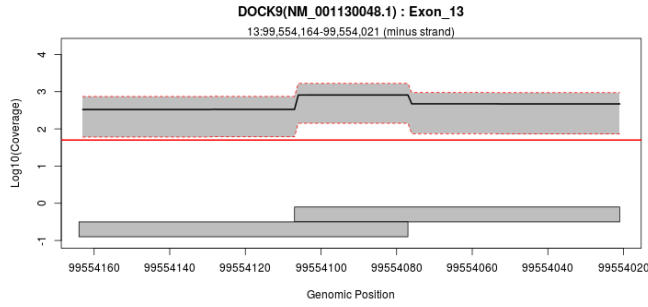
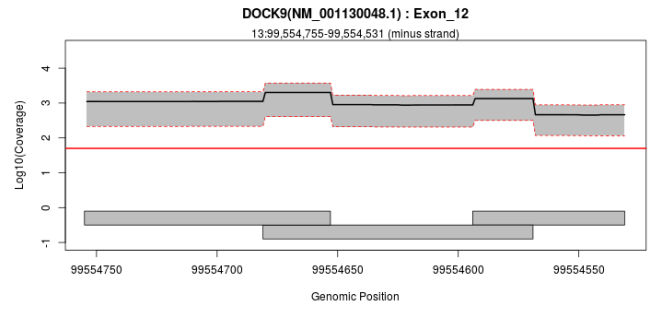
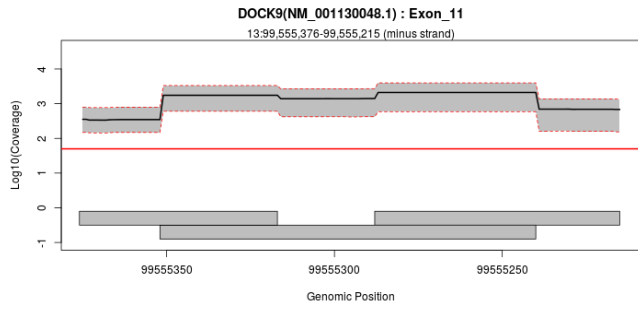


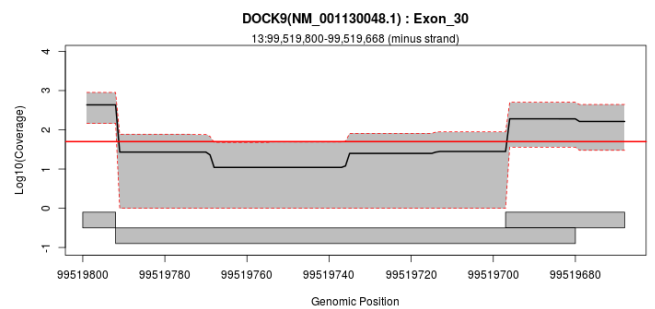
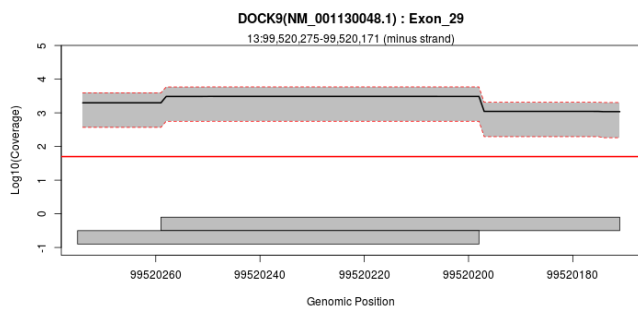
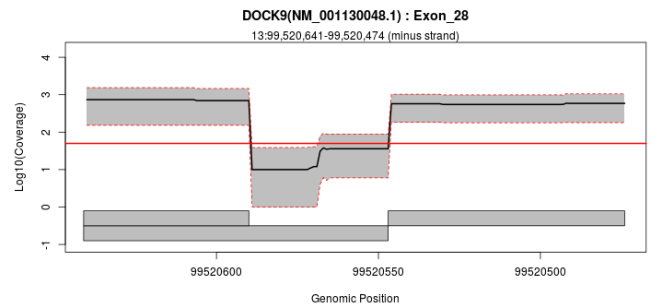
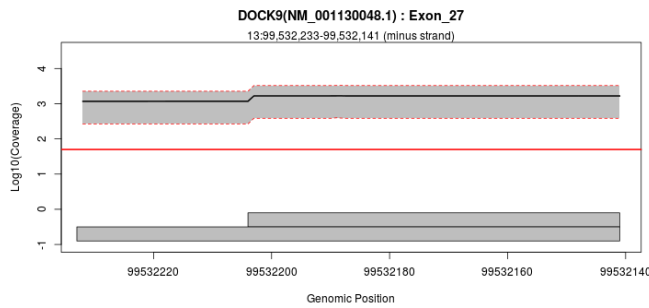
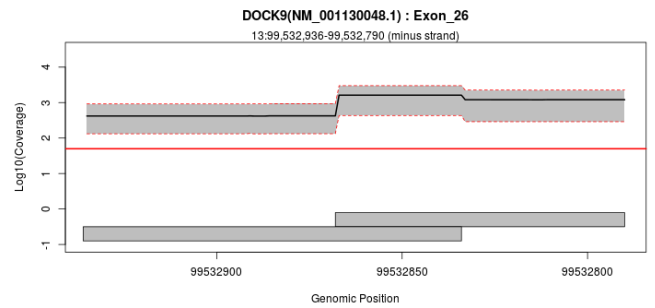
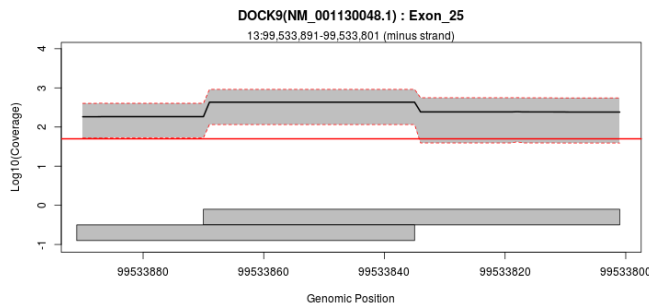
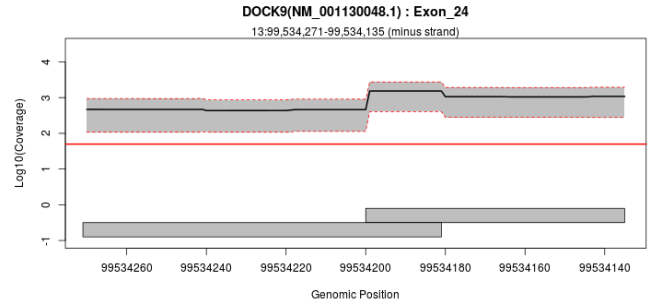
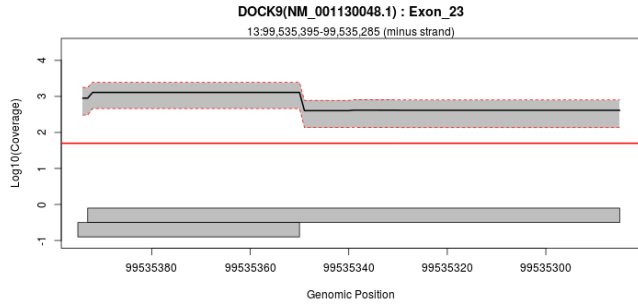
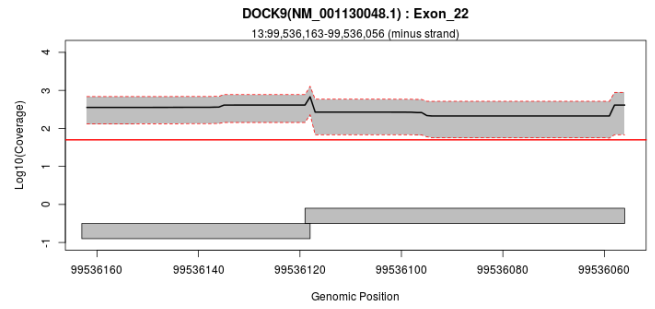
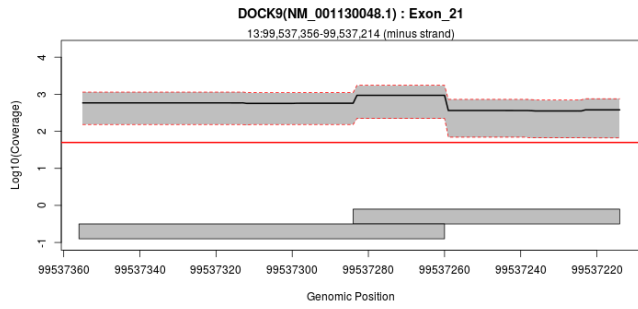


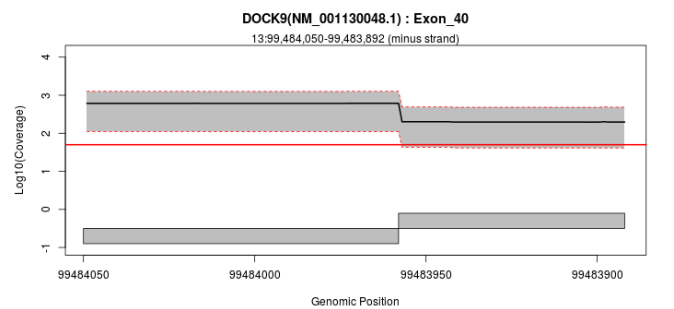
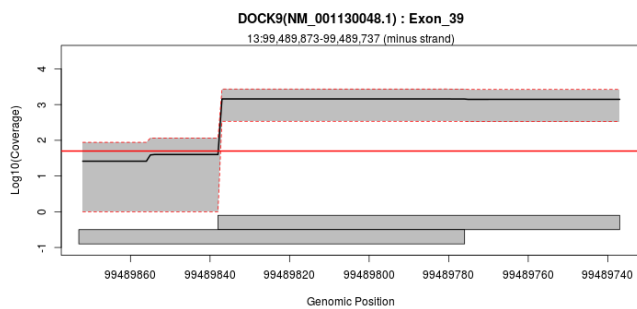
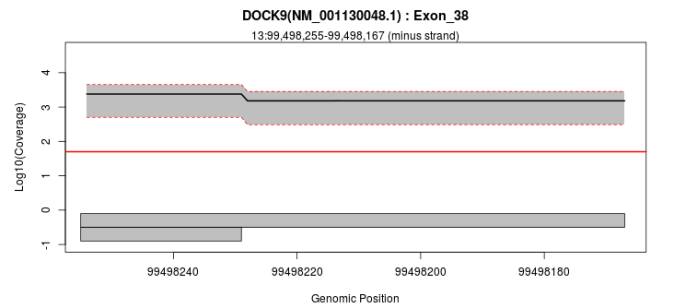
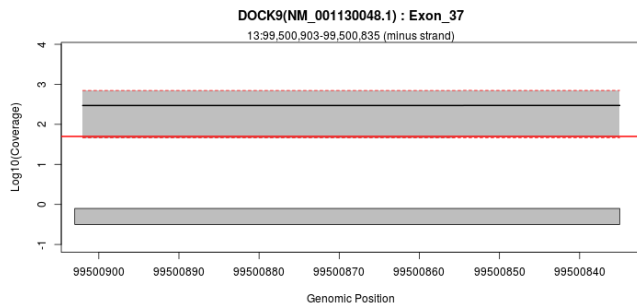
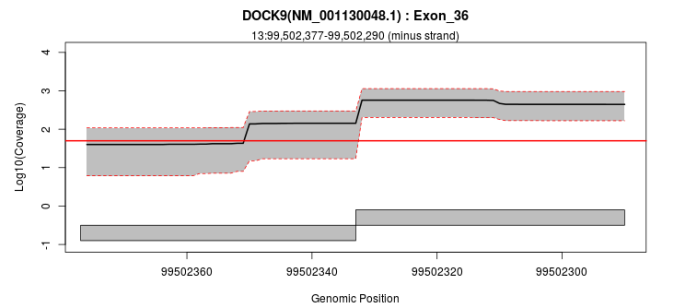
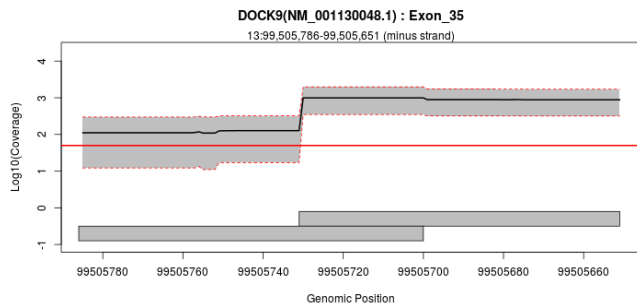
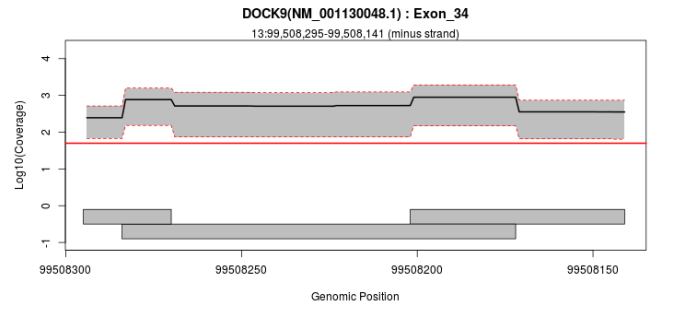
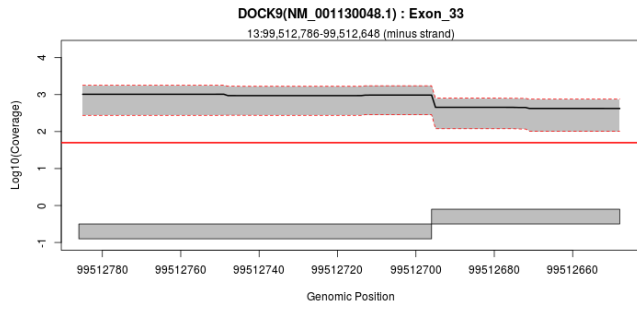
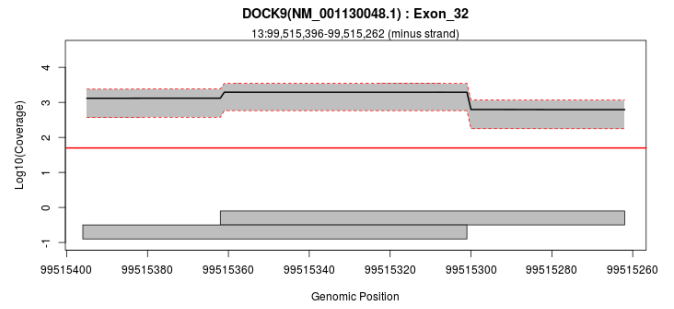
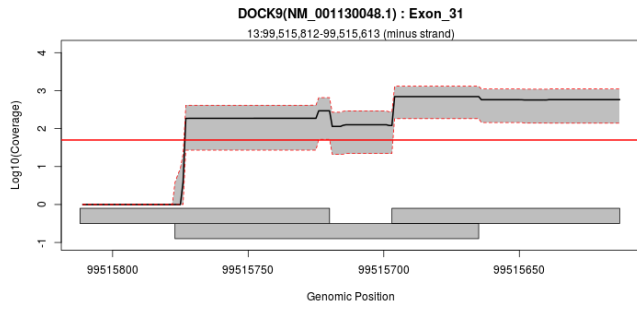


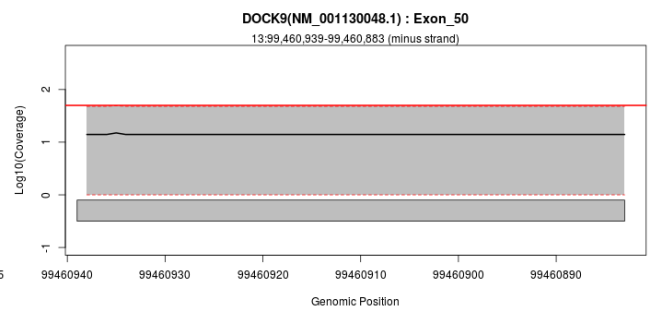
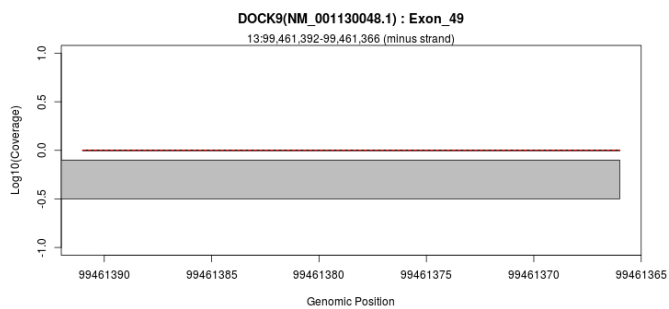
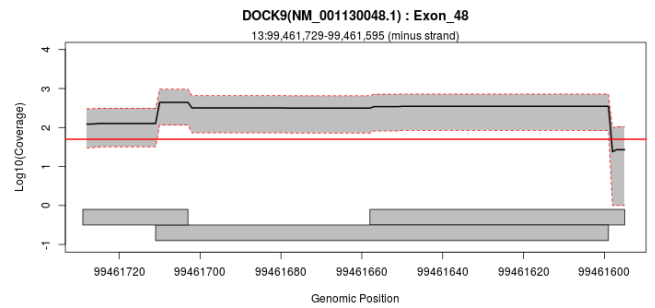
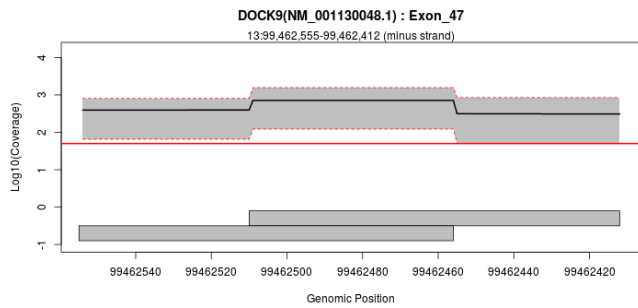
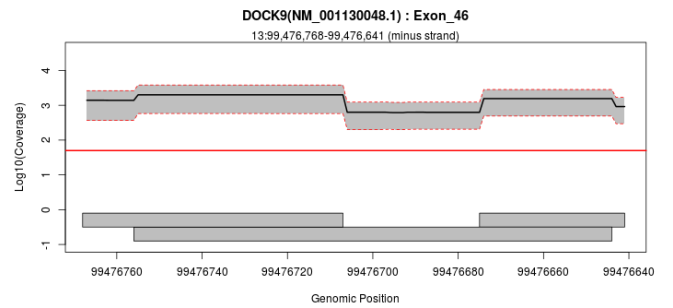
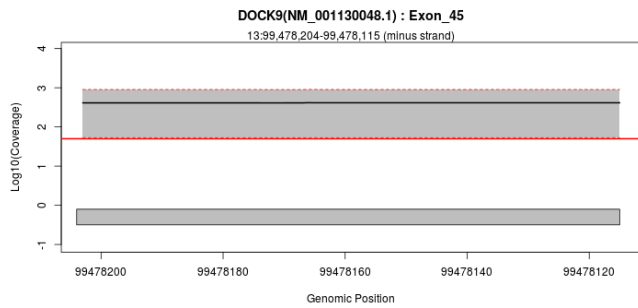
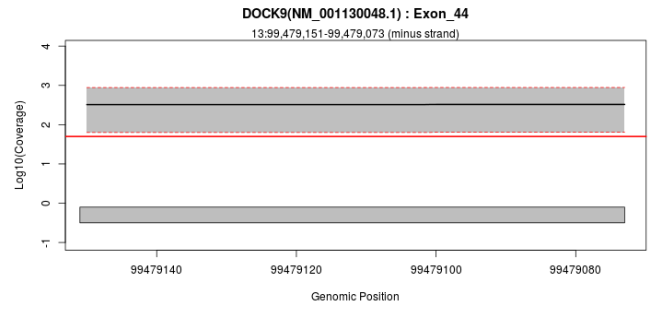
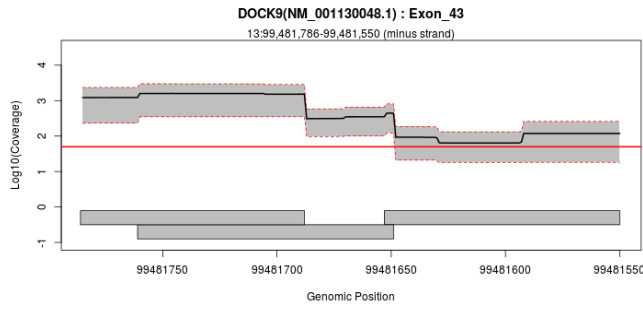
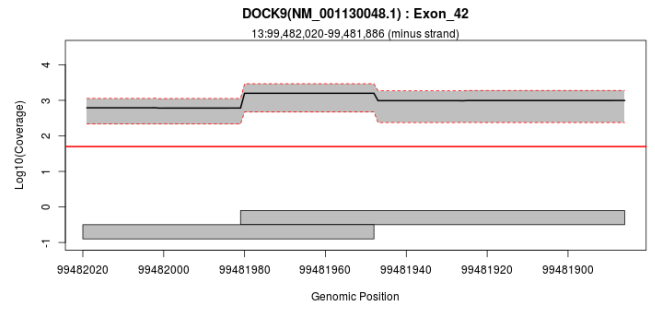
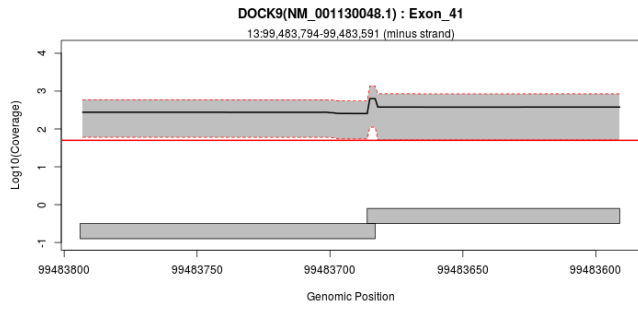
# Exon Overview: DOCK9 : NM\_001130048.1

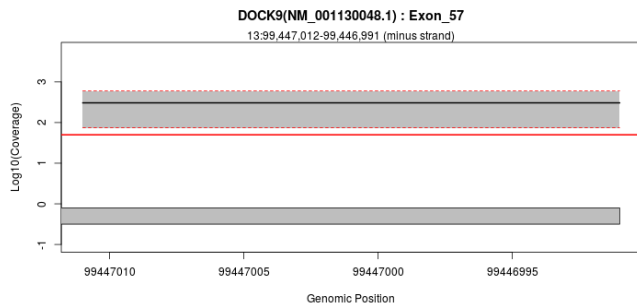
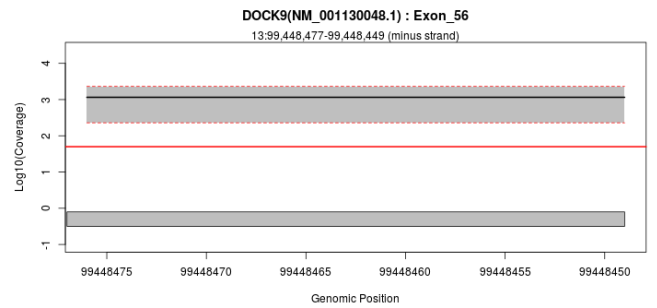
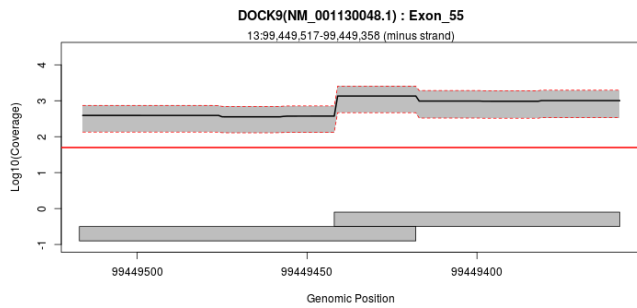
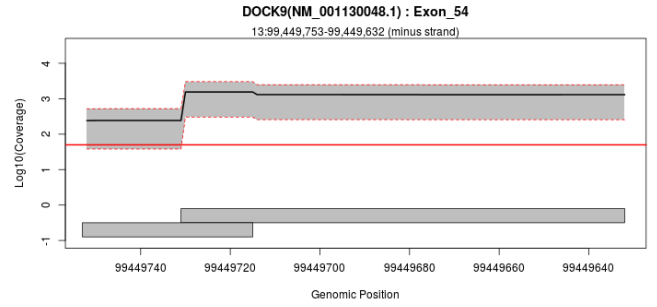
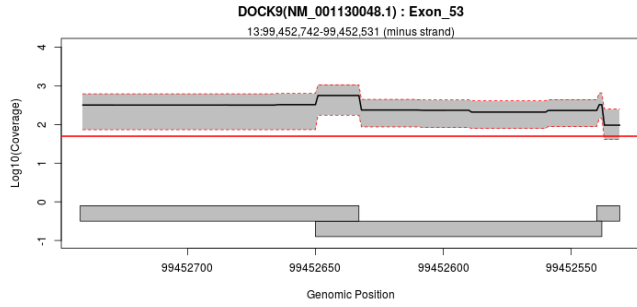
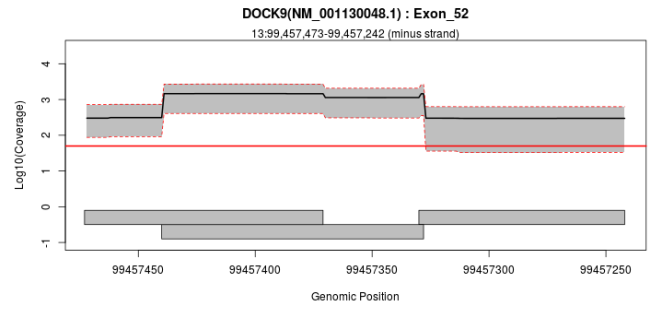
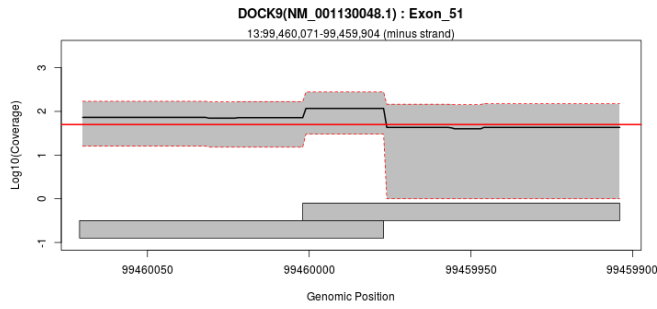




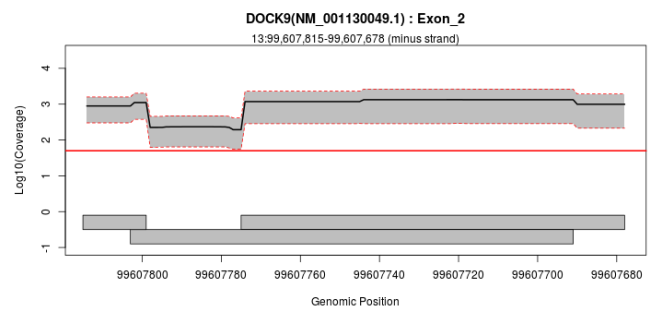
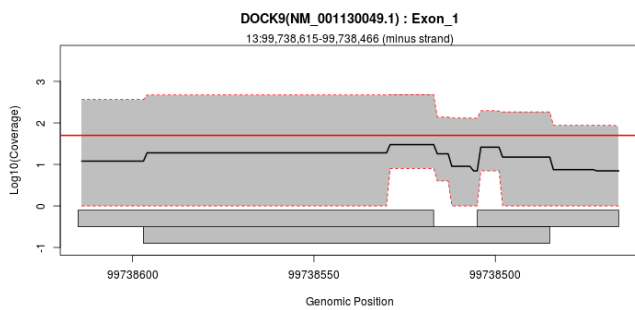


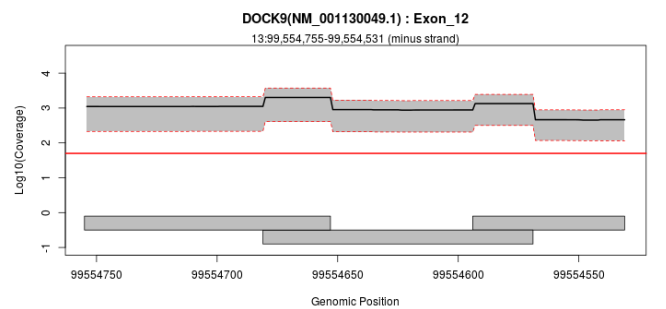
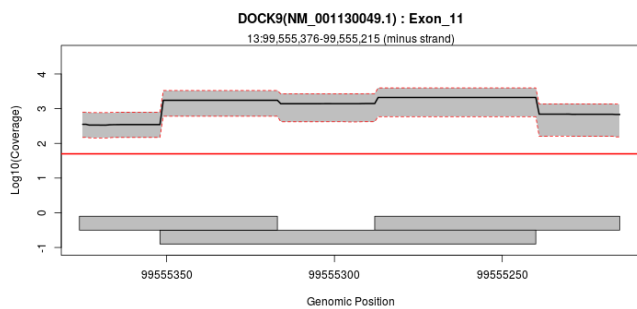
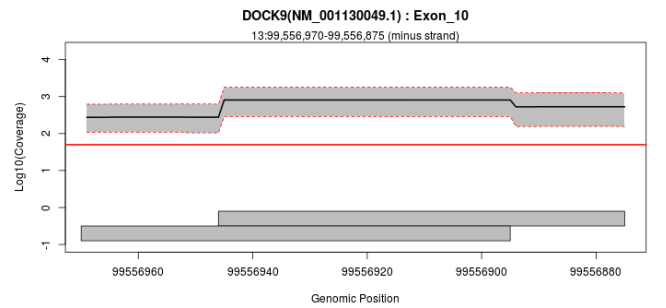
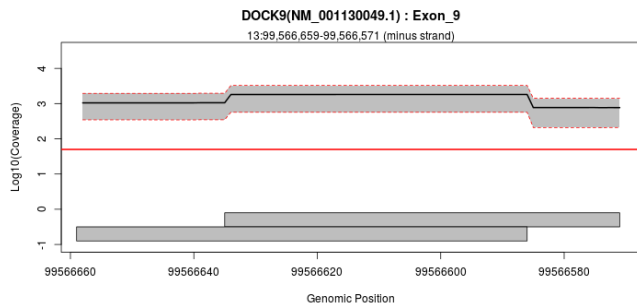
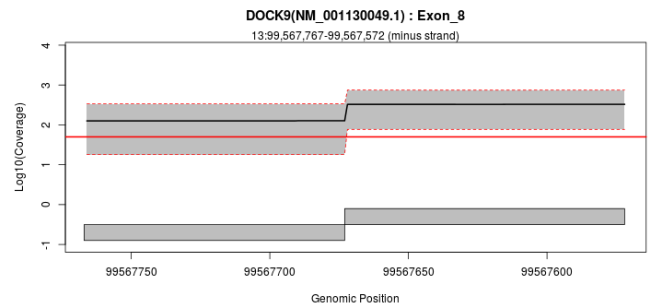
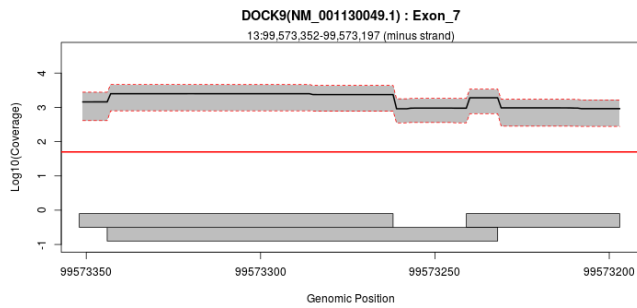
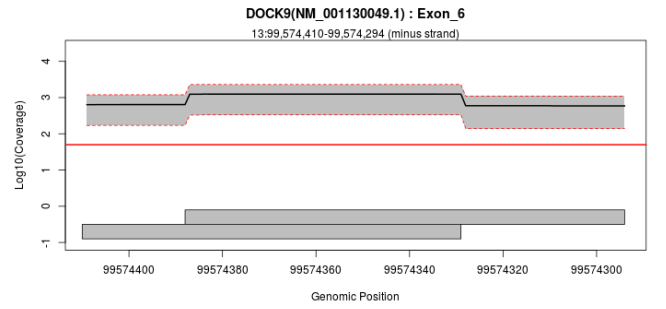
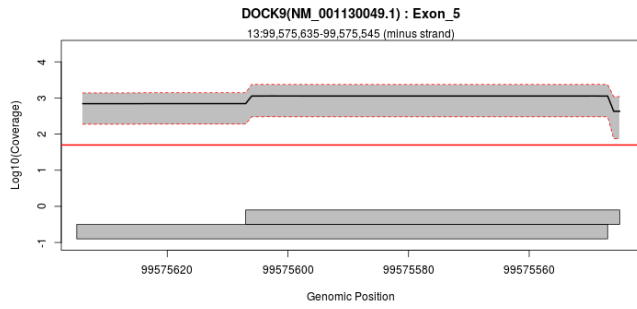
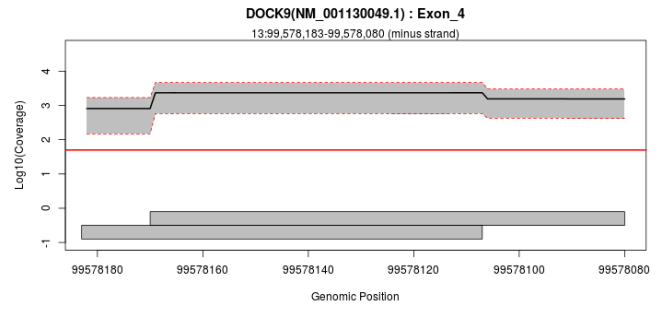
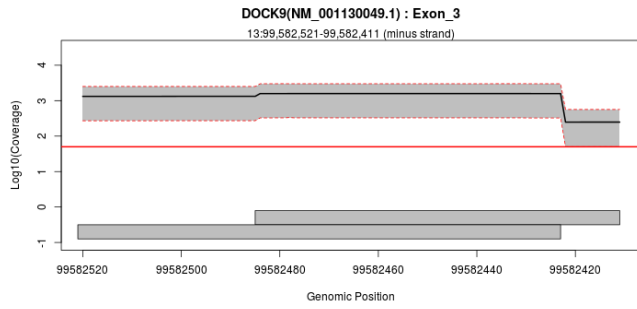




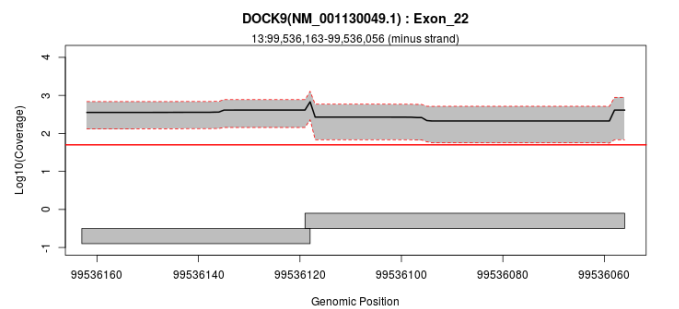
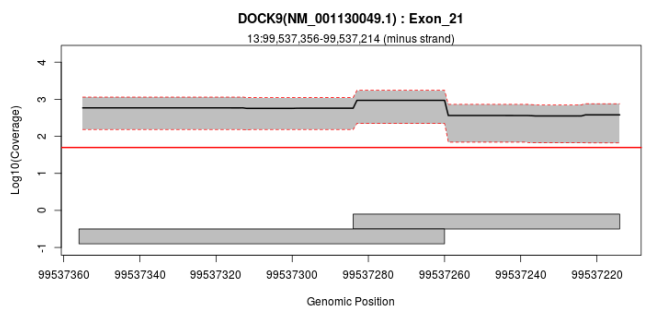
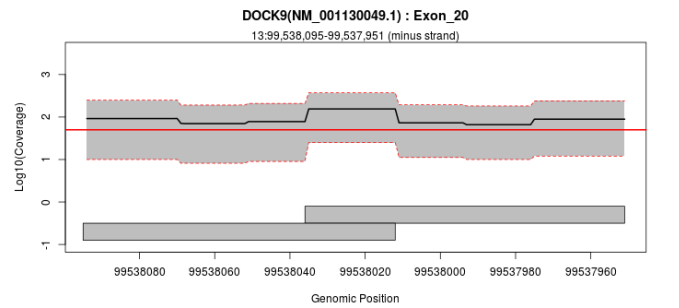
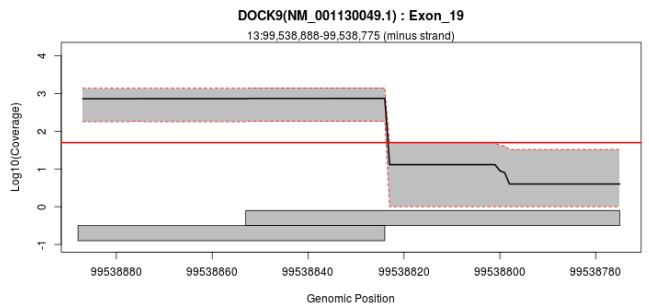
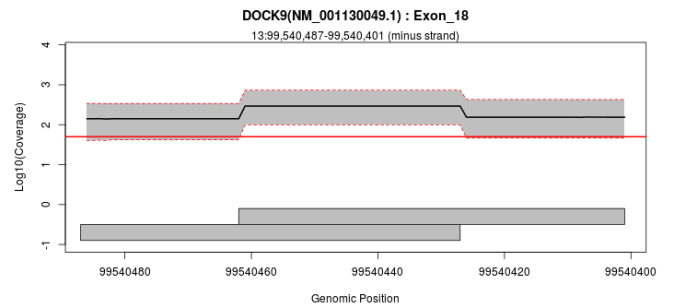
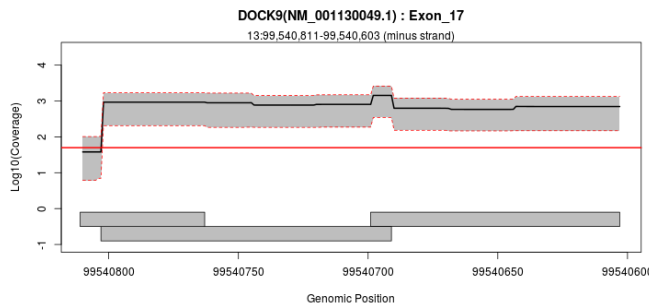
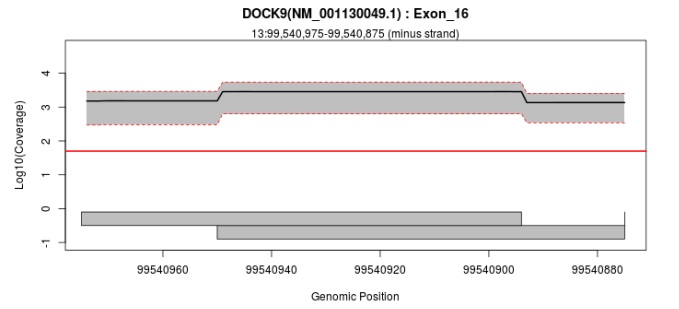
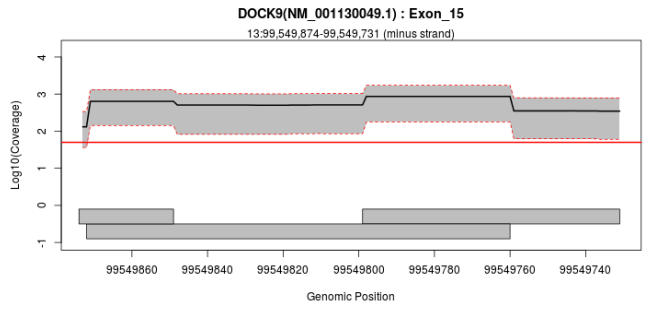
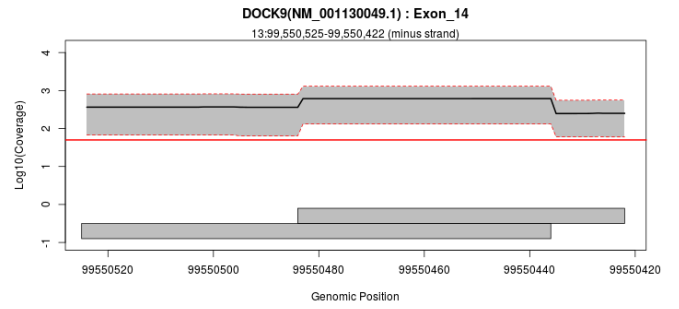
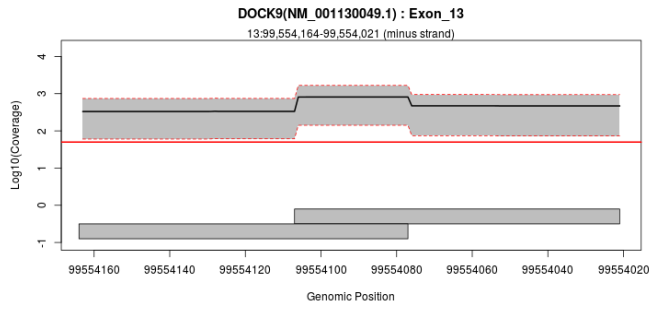


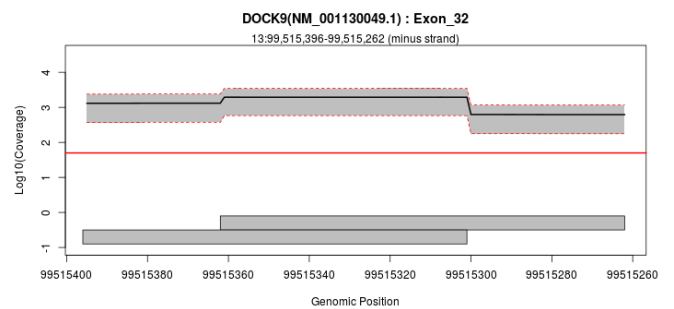
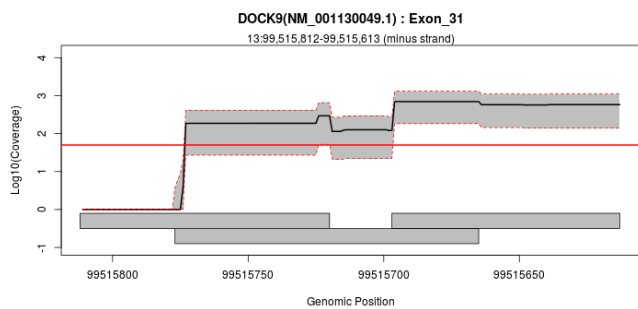
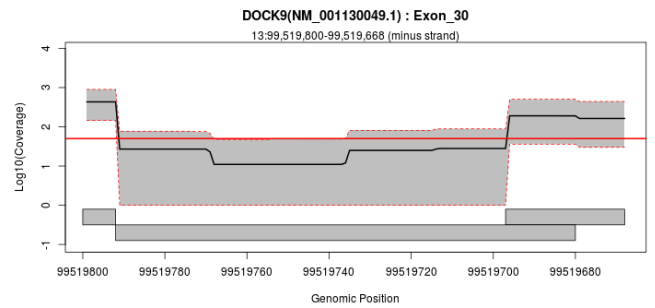
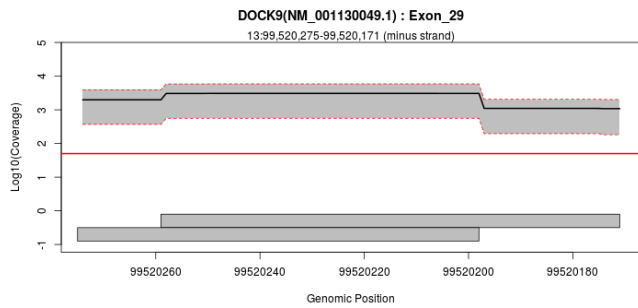
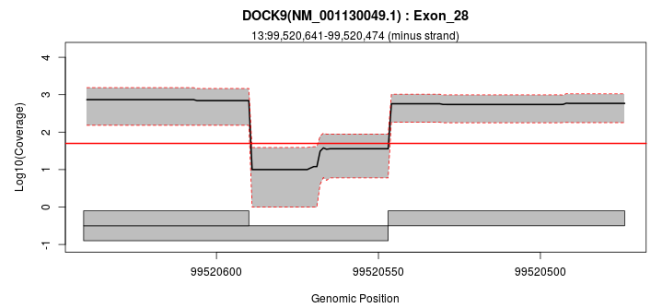
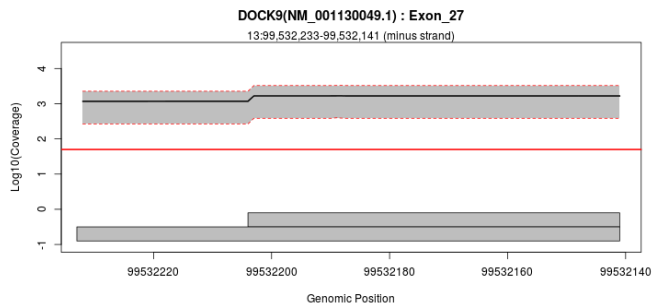
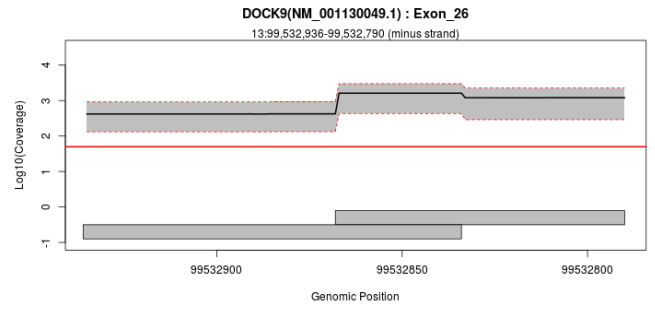
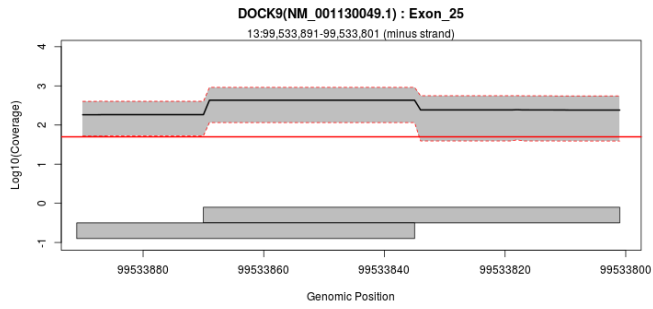
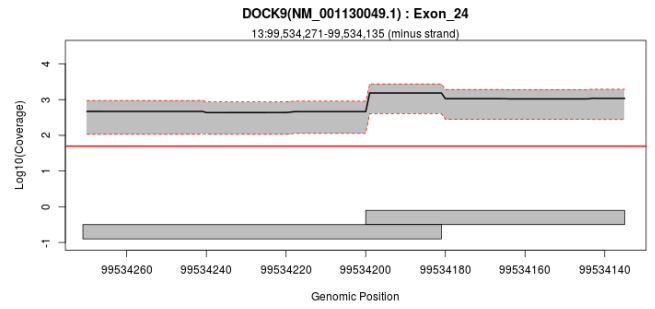
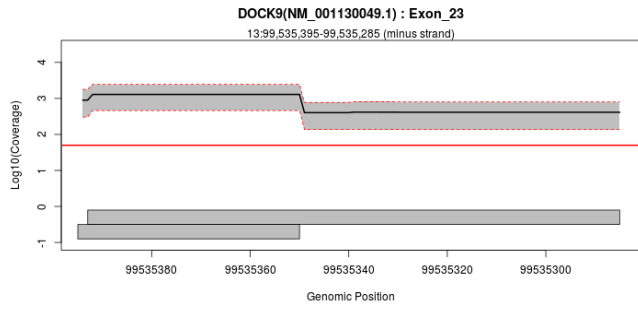
## Exon Overview: DOCK9 : NM\_001130049.1

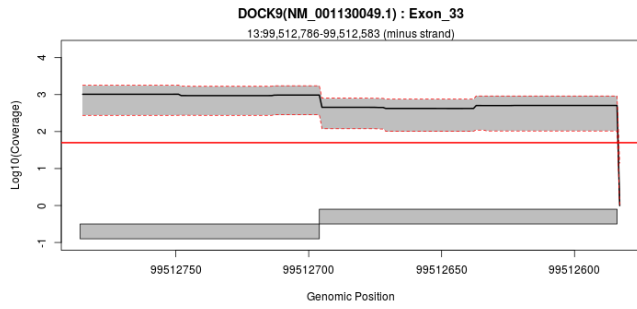




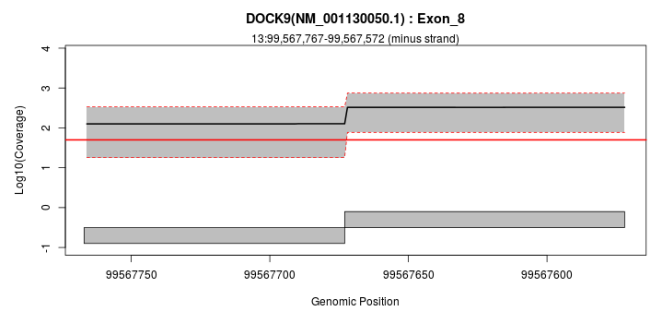
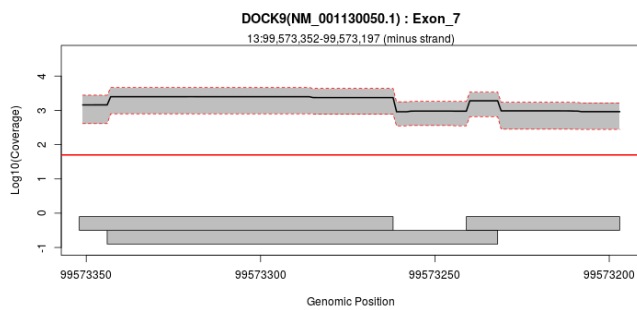
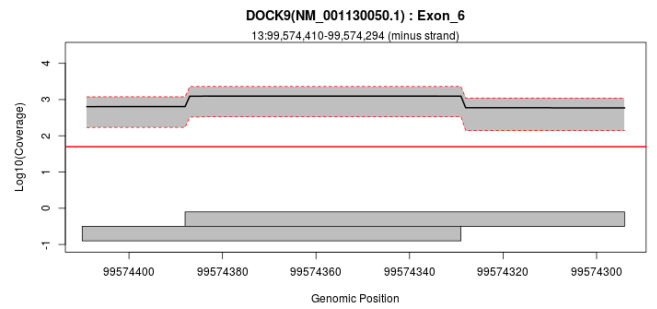
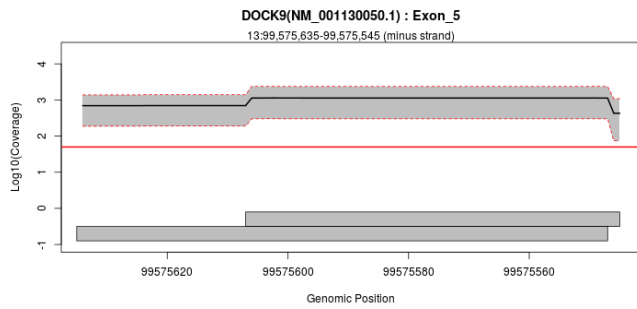
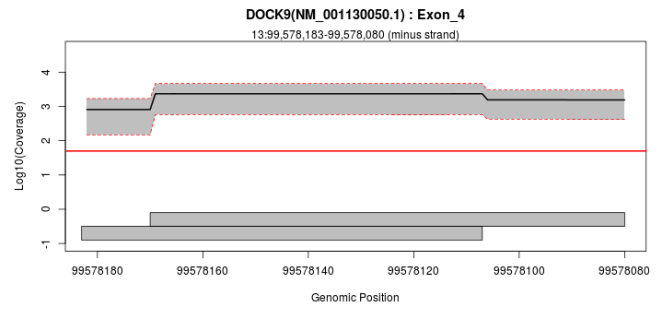
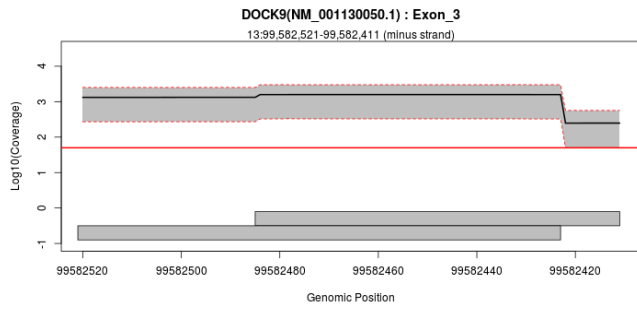
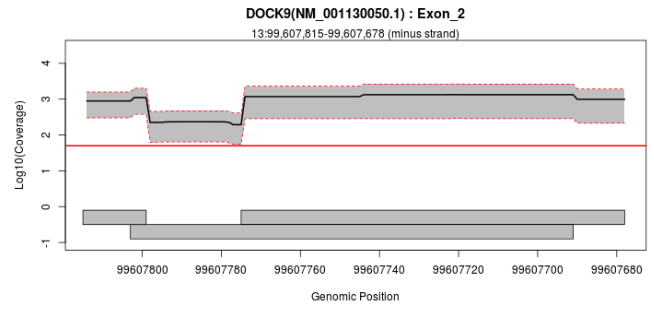
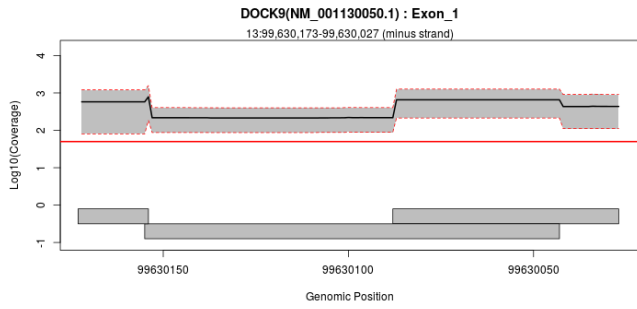


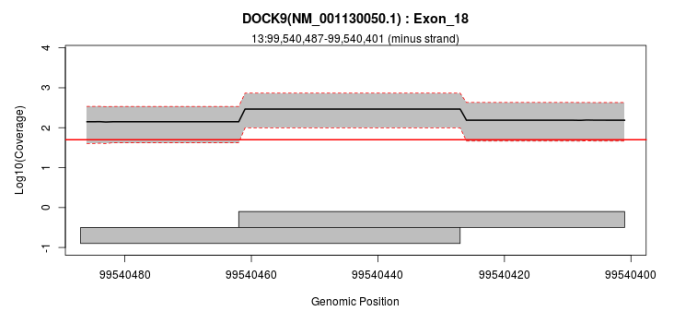
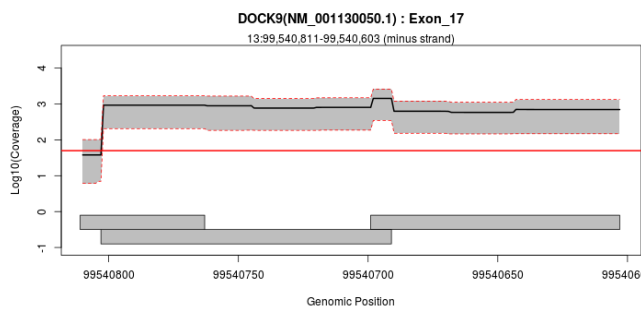
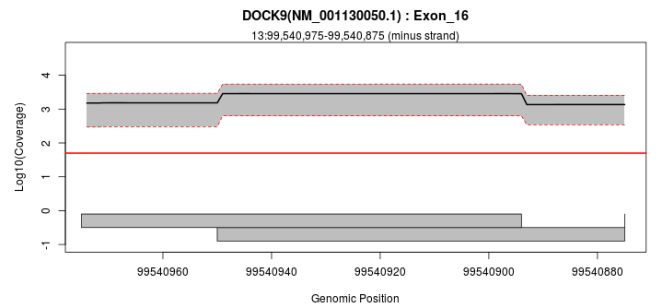
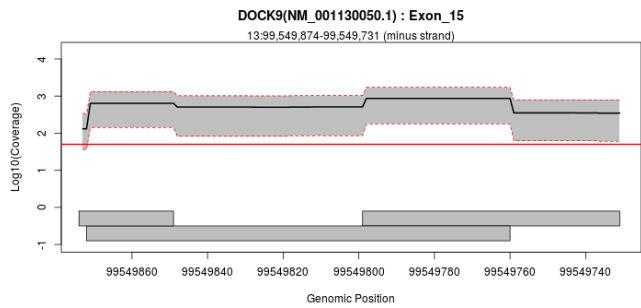
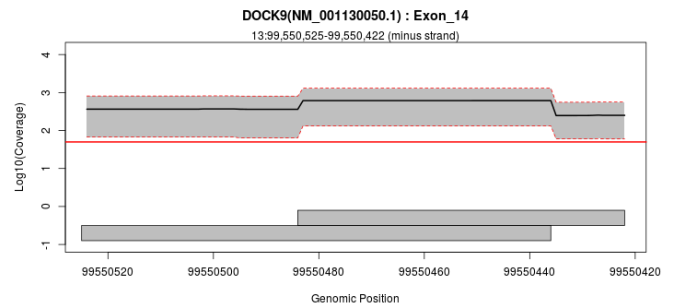
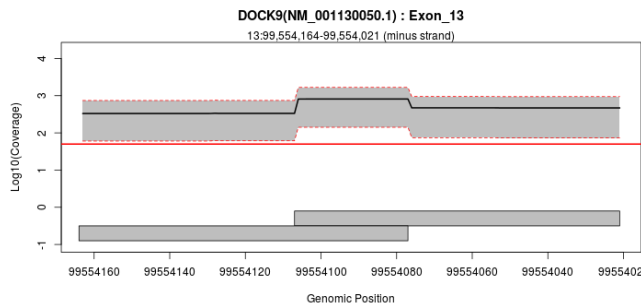
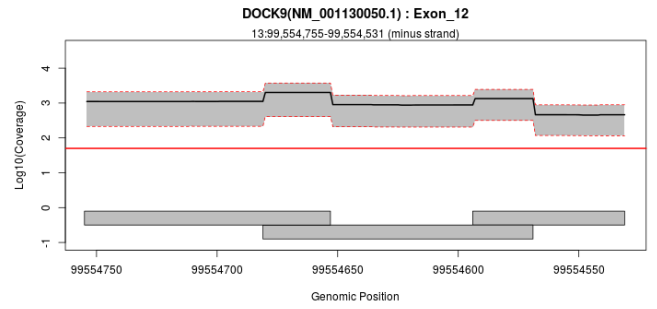
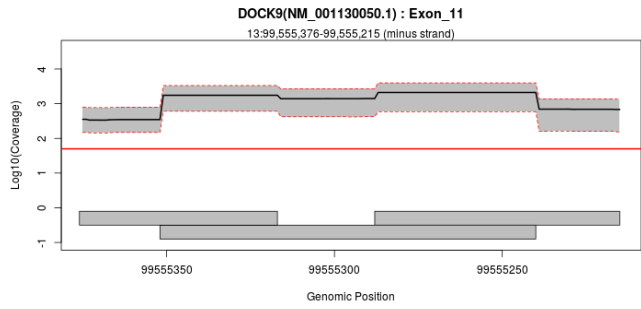
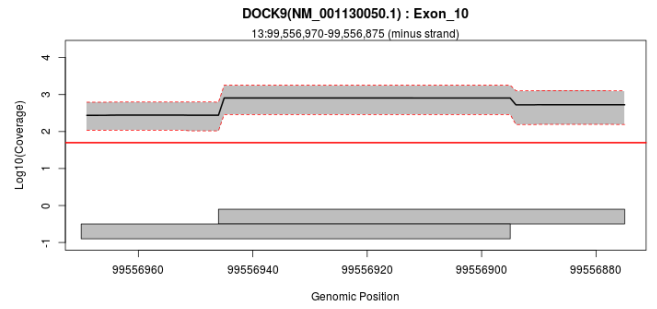
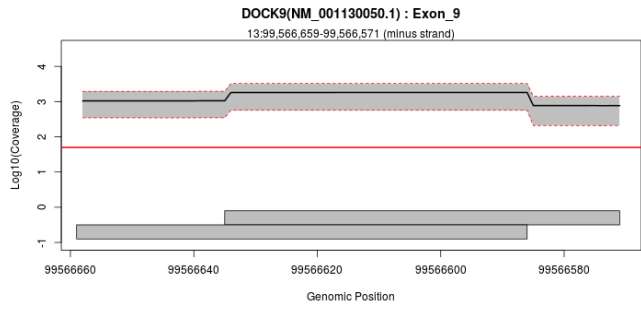


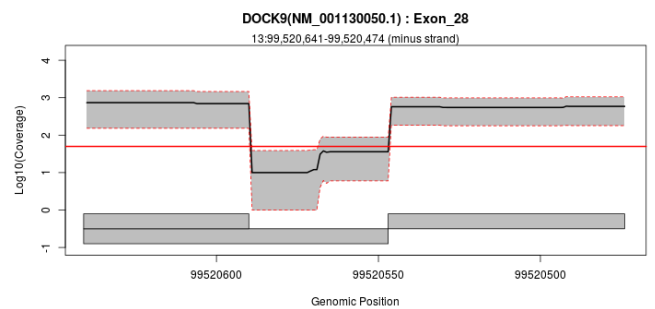
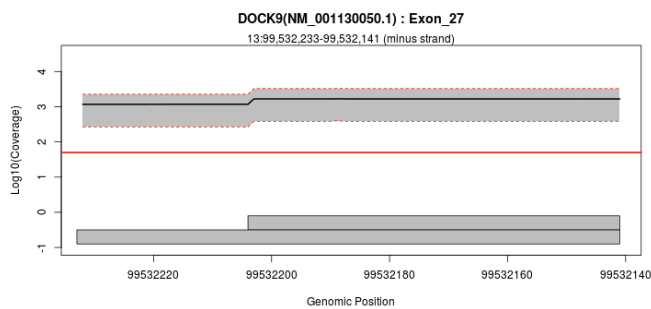
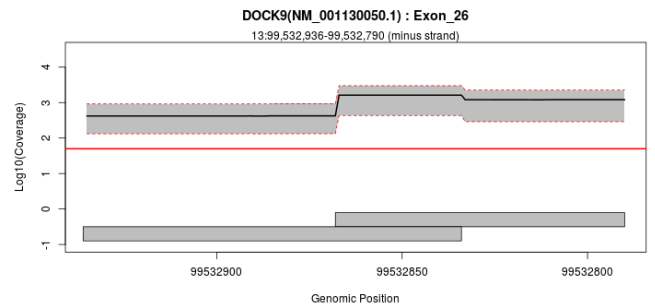
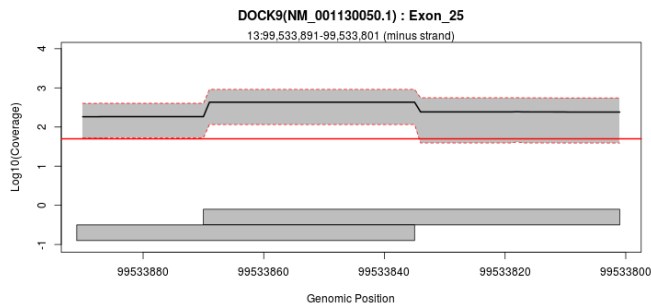
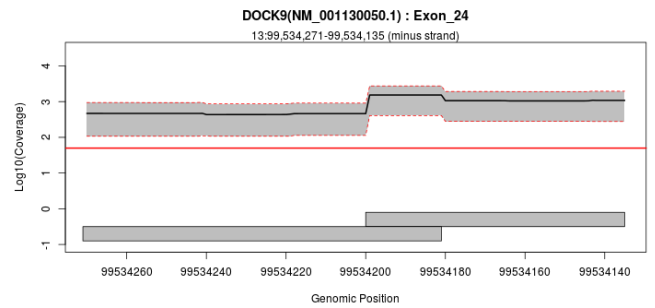
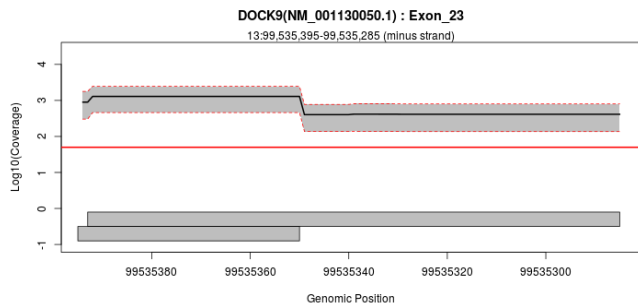
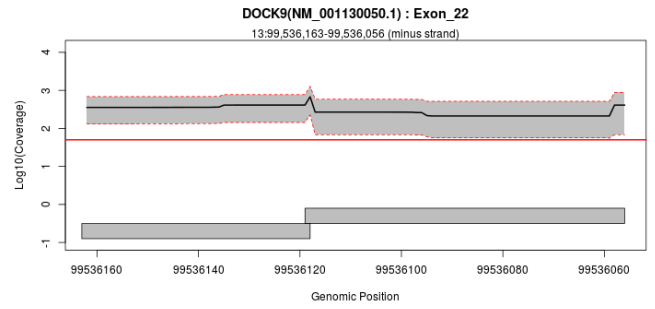
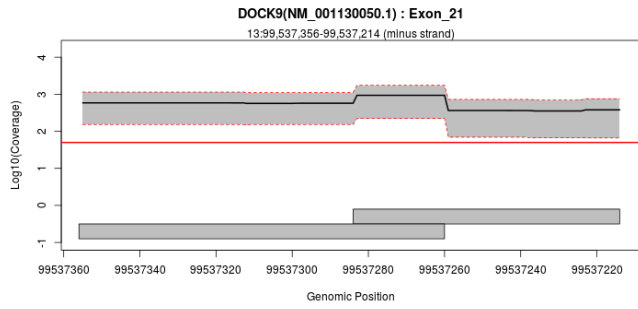
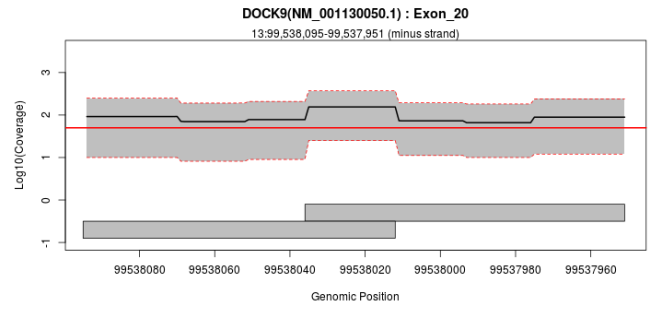
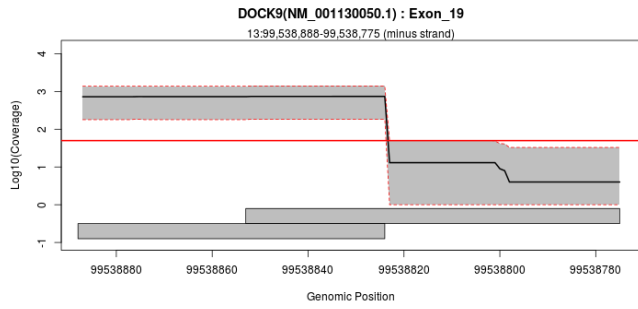


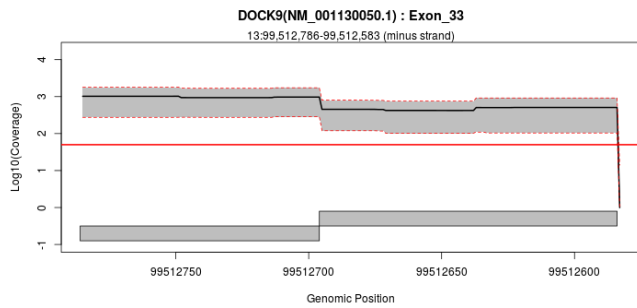
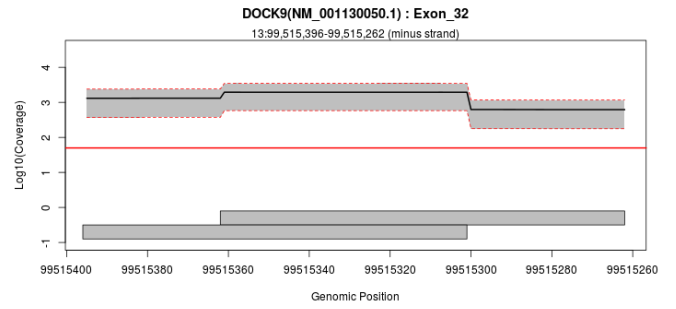
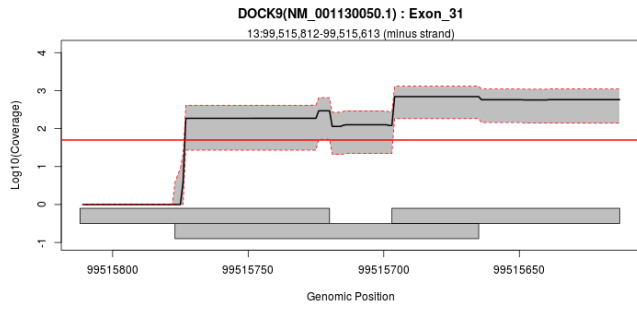
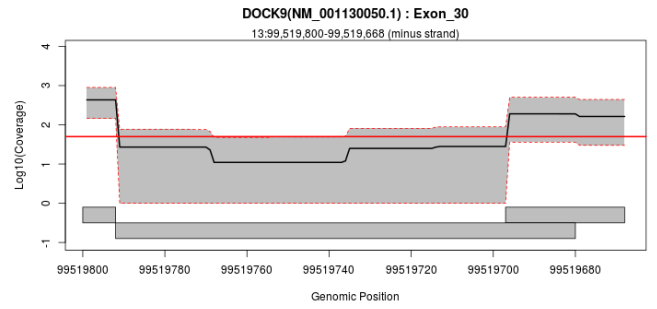
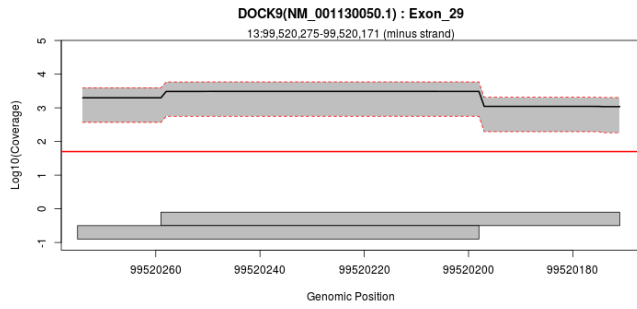


## Exon Overview: DOCK9 : NM\_001130050.1

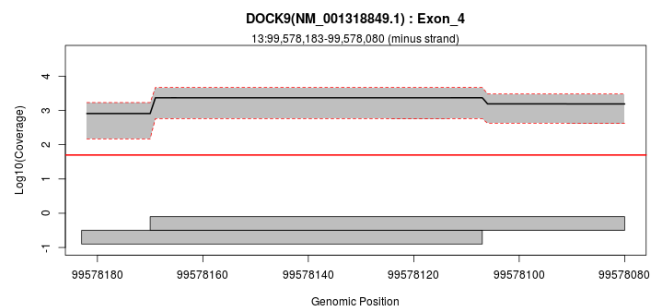
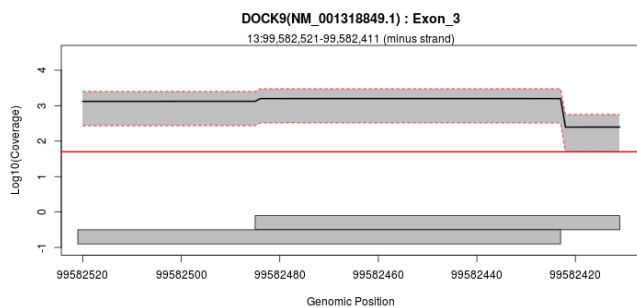
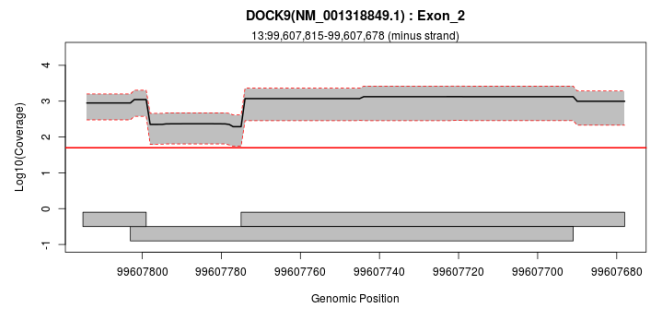
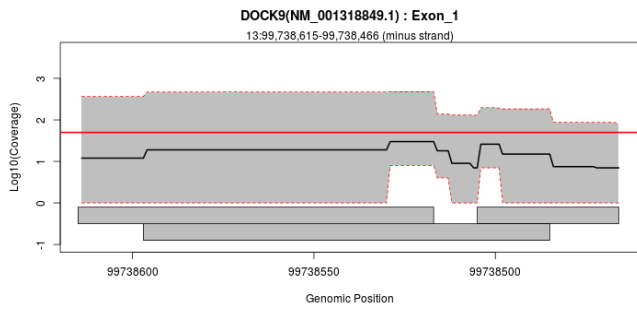


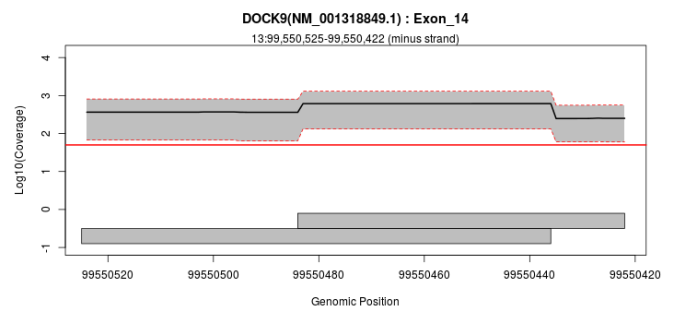
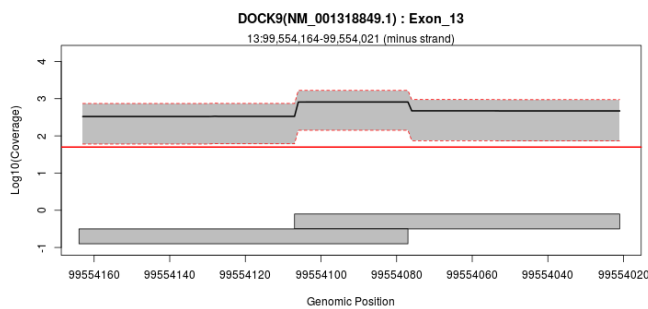
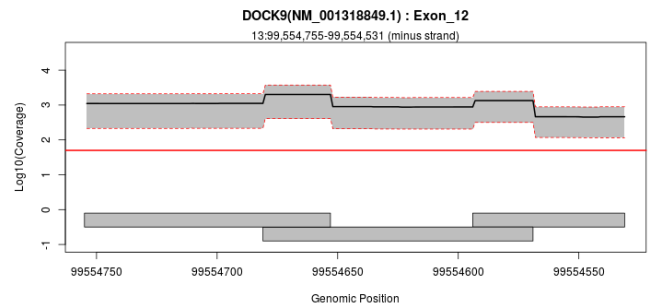
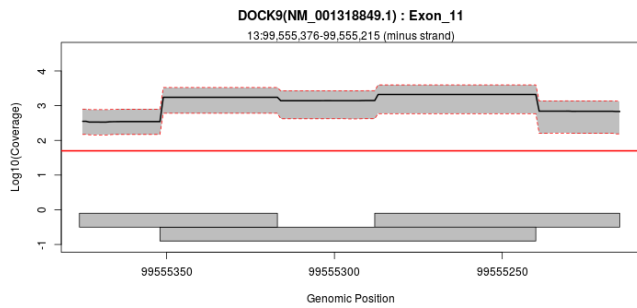
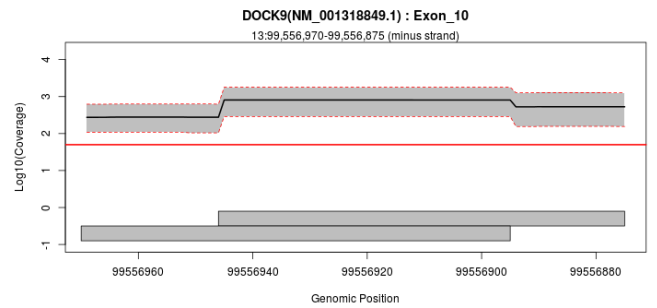
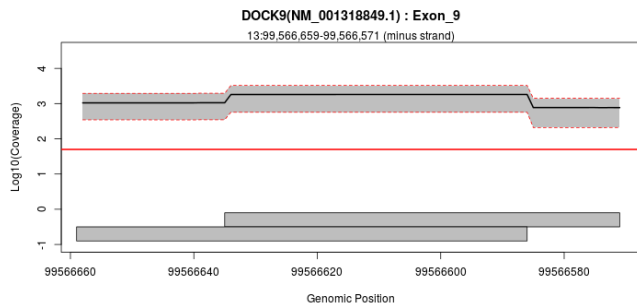
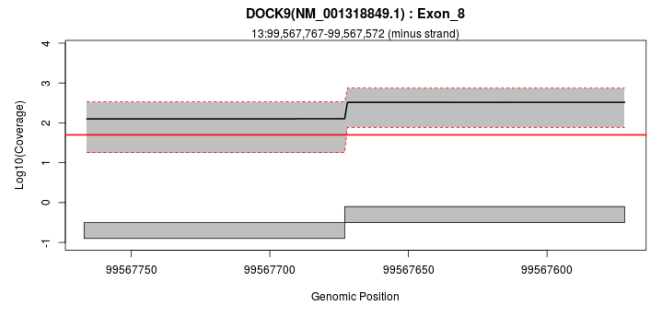
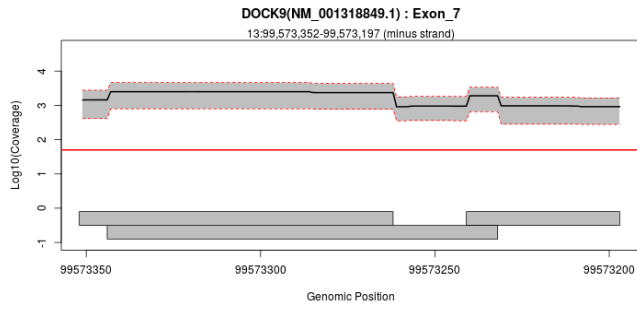
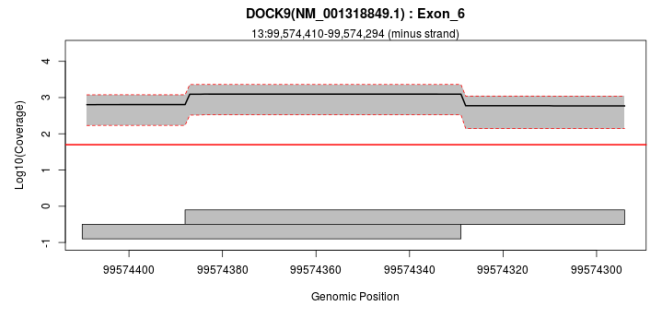
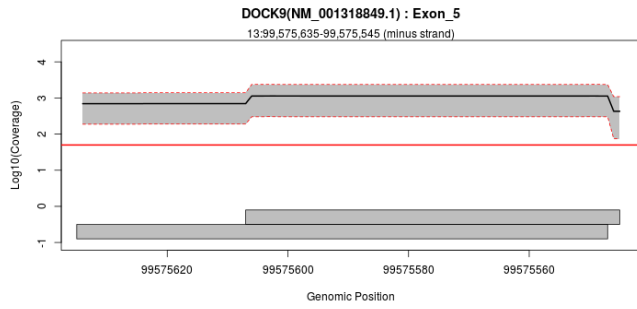


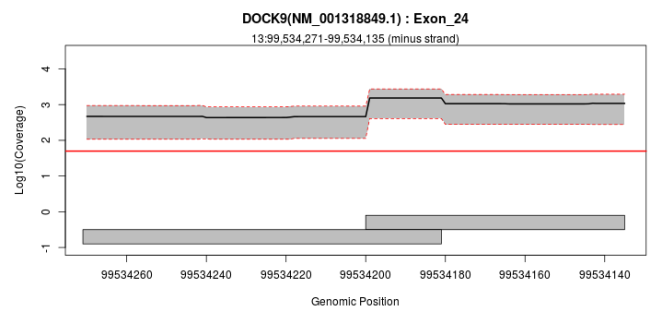
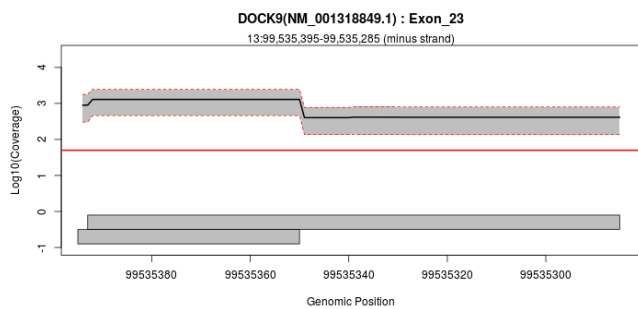
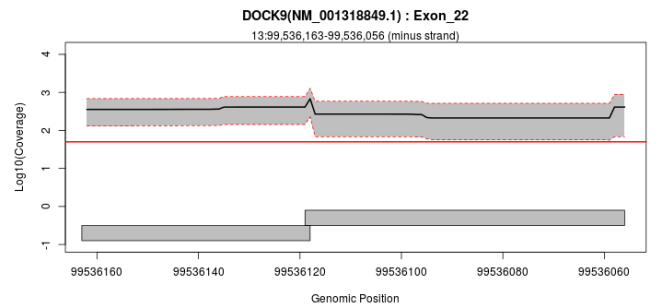
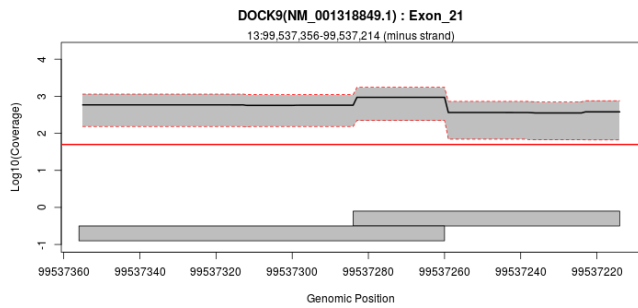
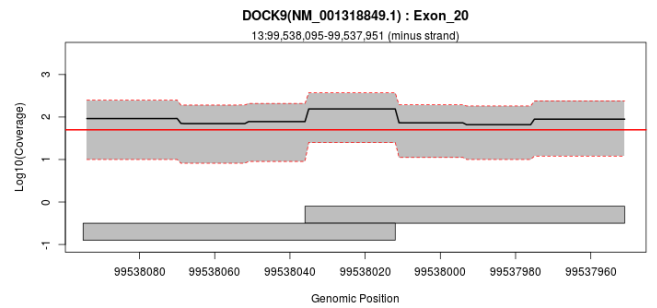
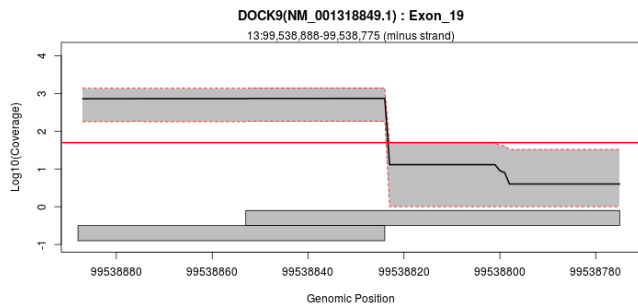
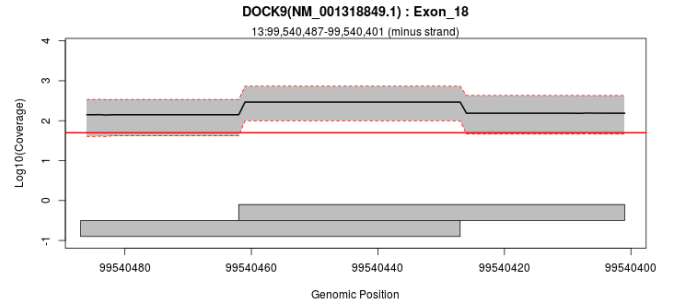
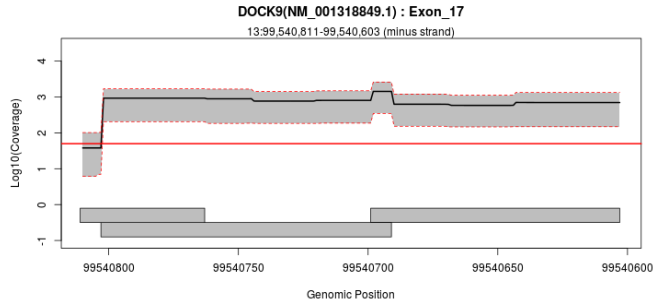
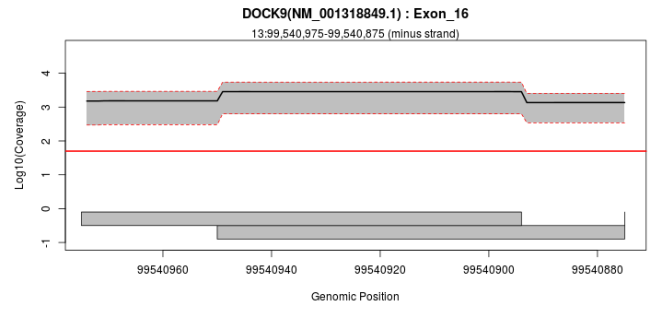
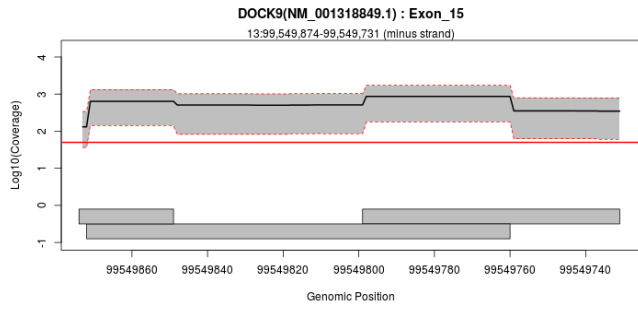




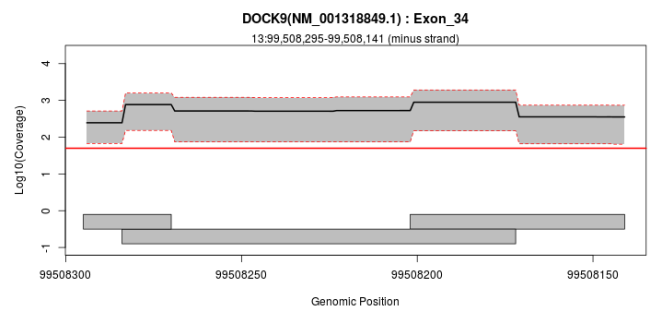
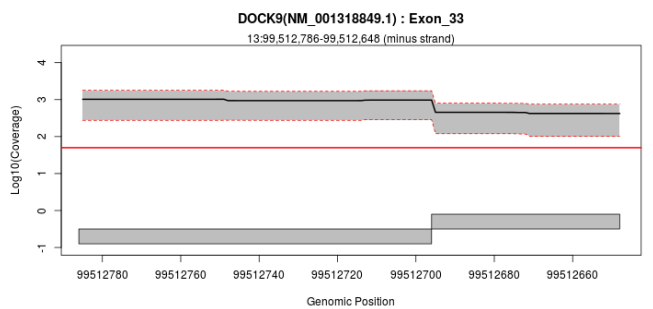
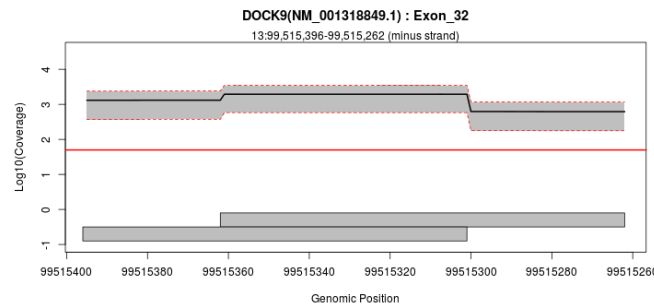
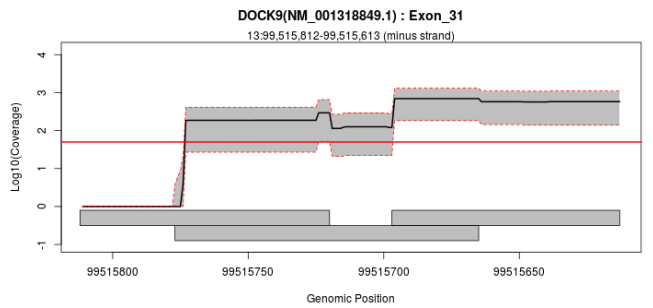
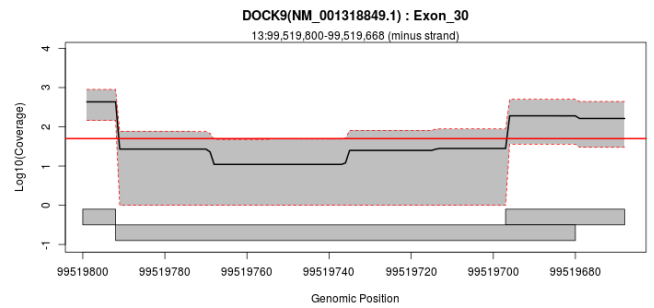
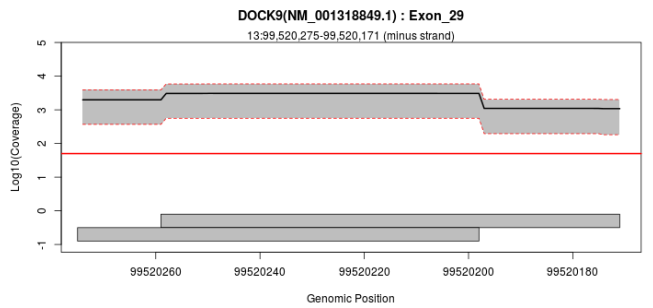
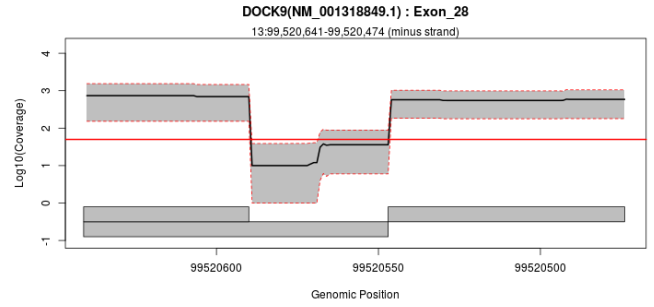
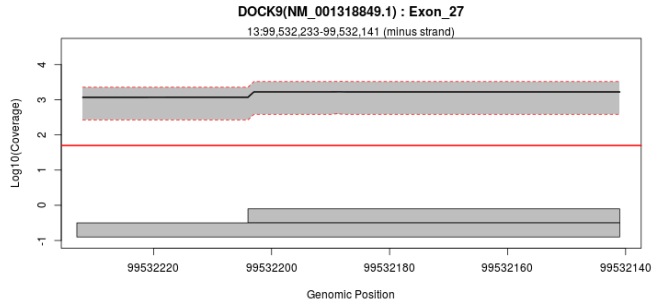
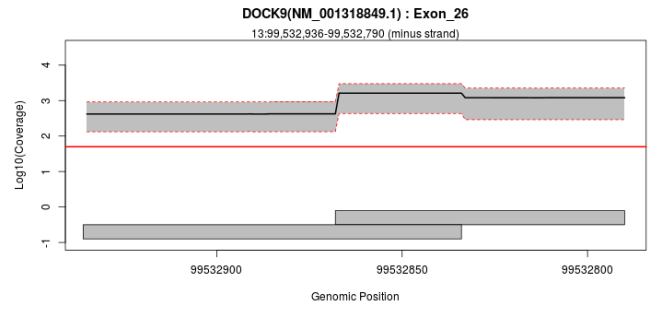
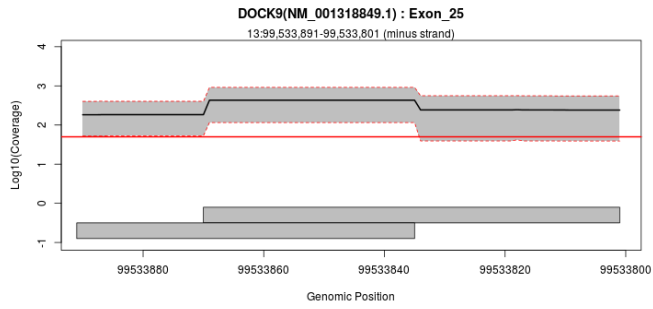
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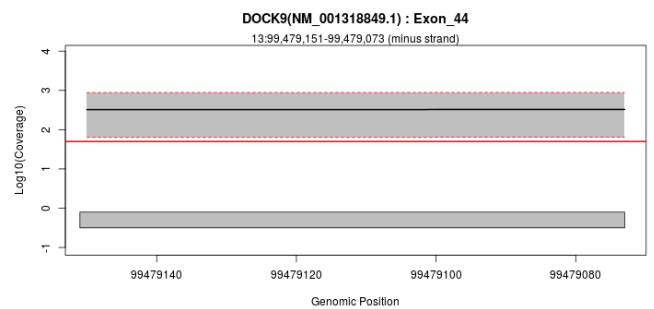
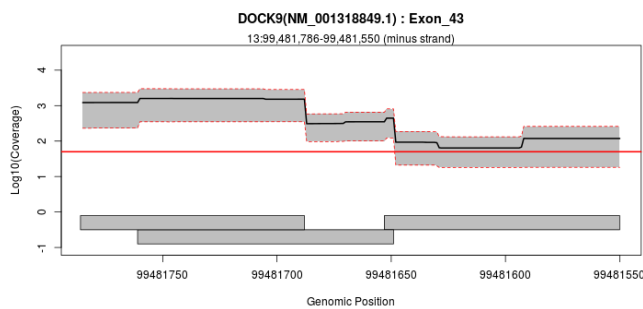
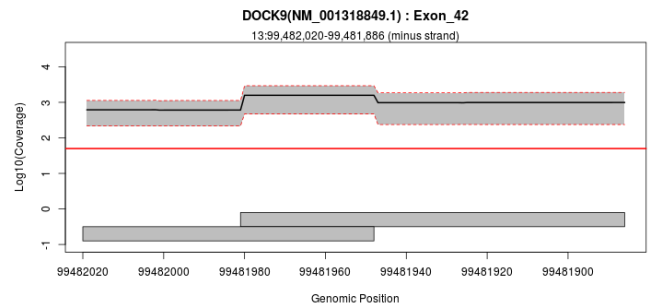
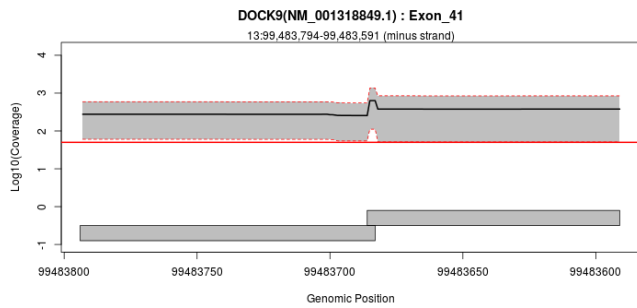
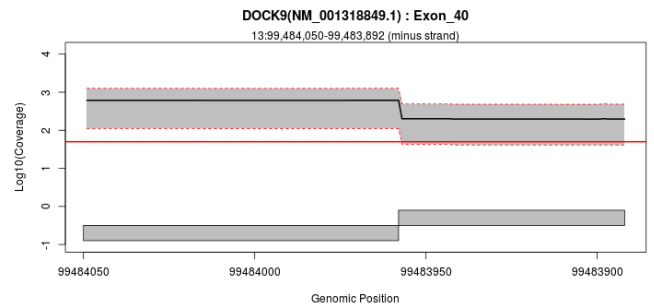
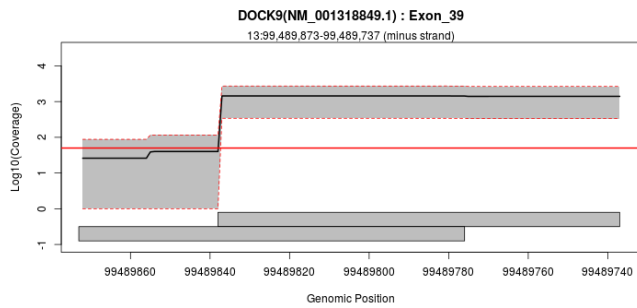
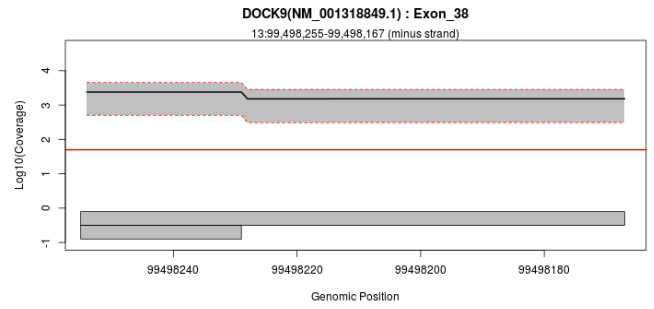
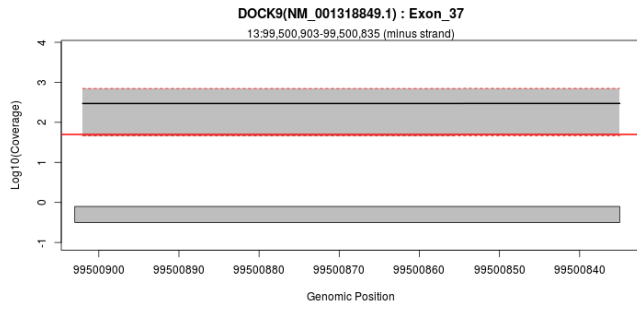
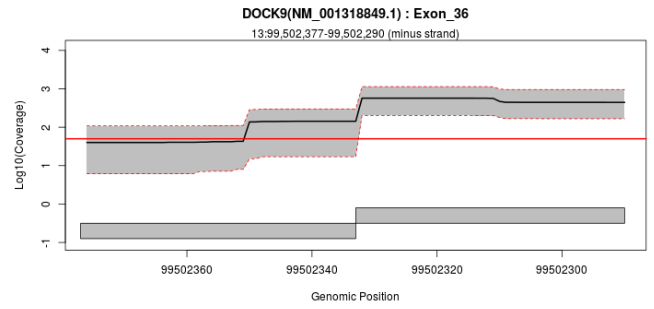
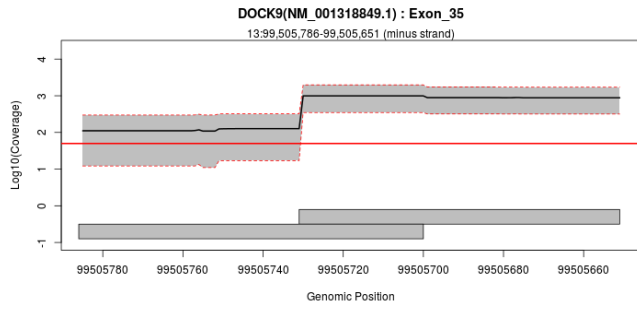


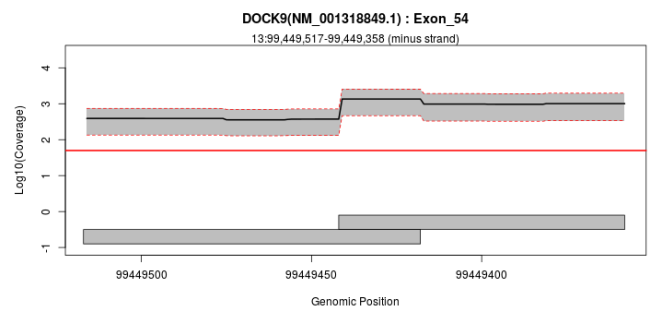
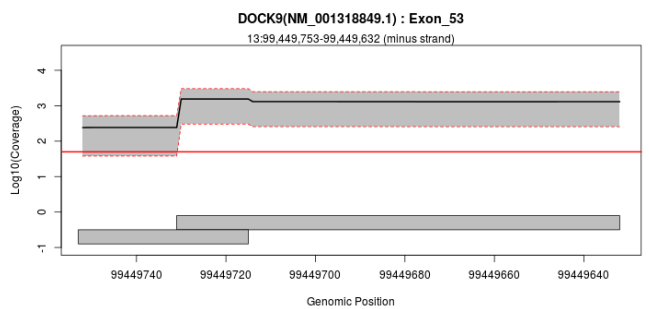
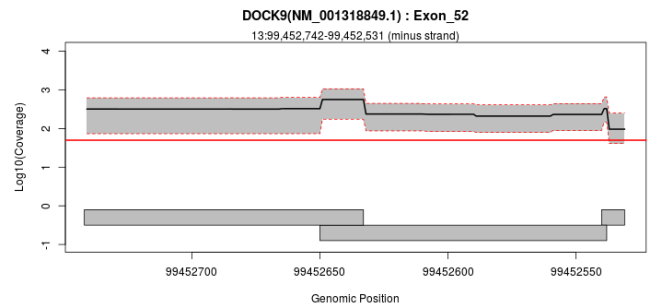
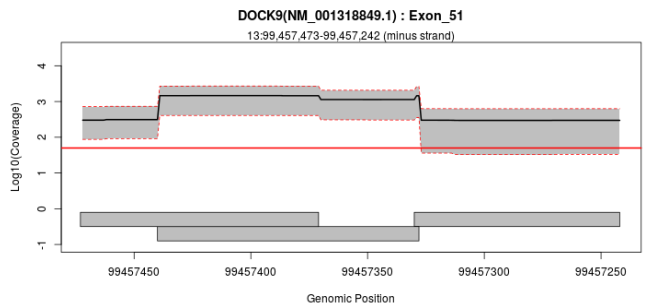
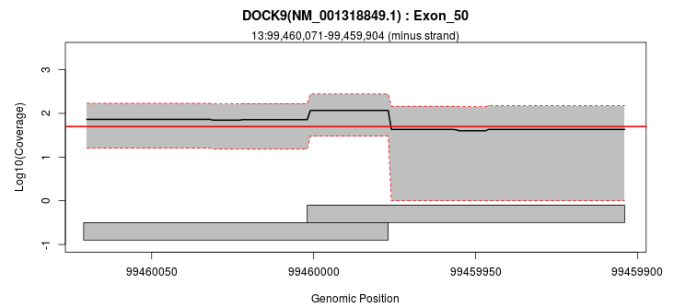
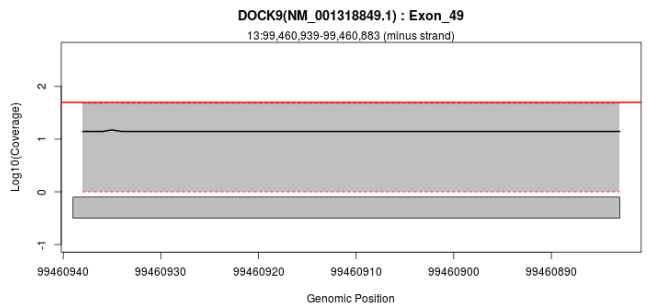
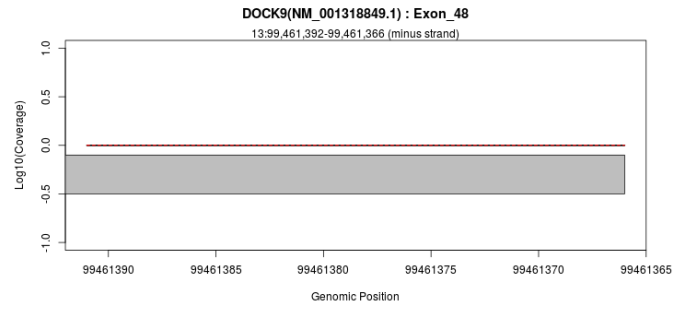
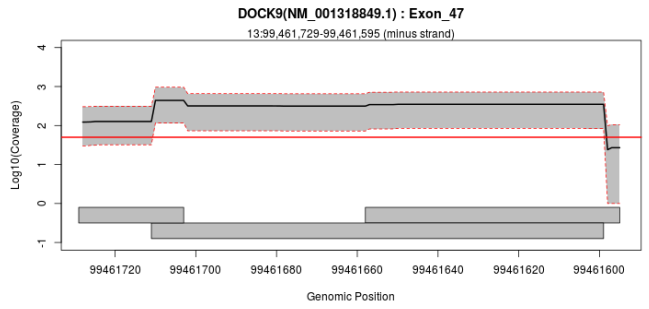
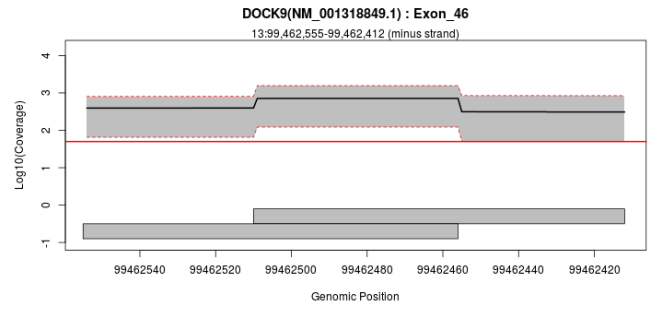
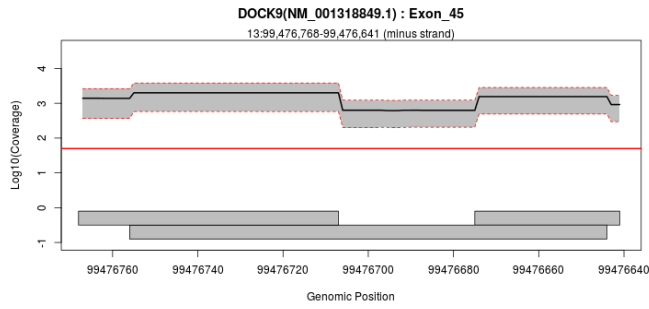


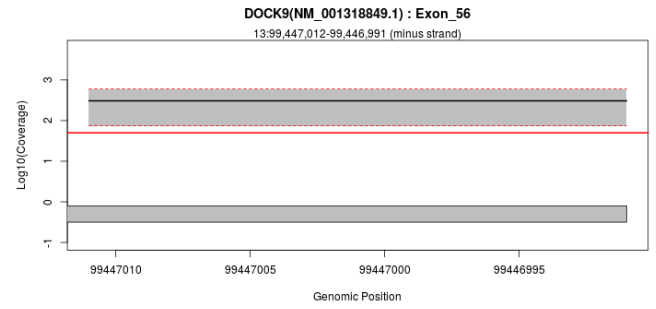
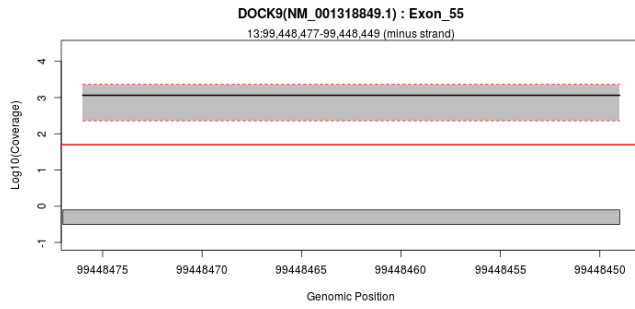




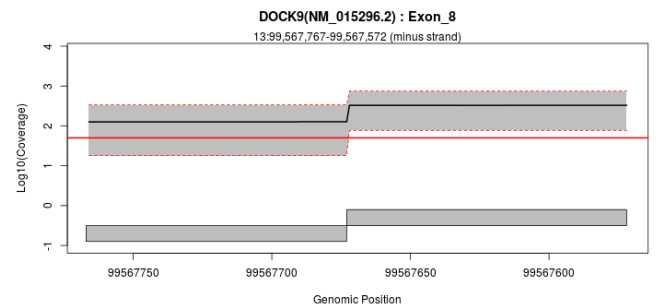
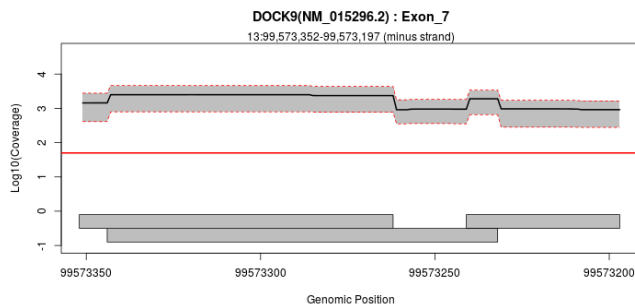
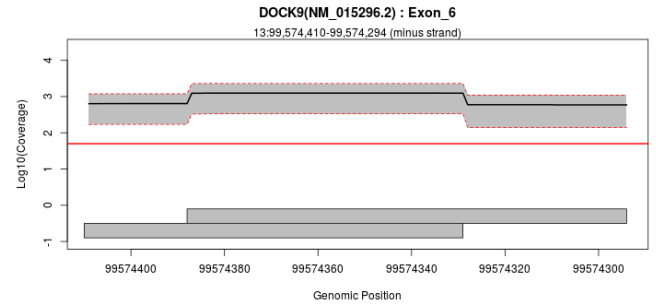
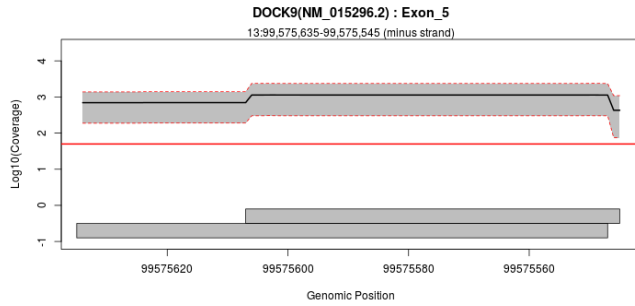
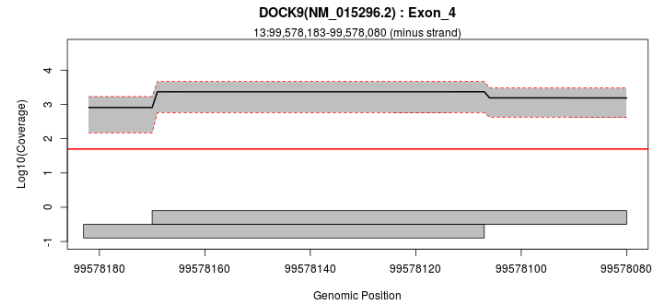
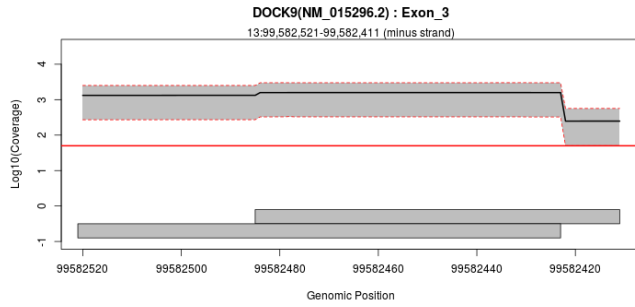
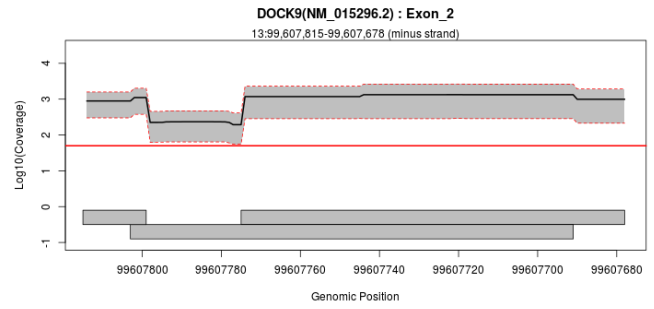
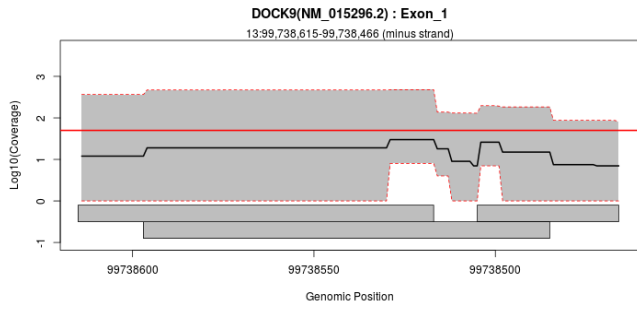


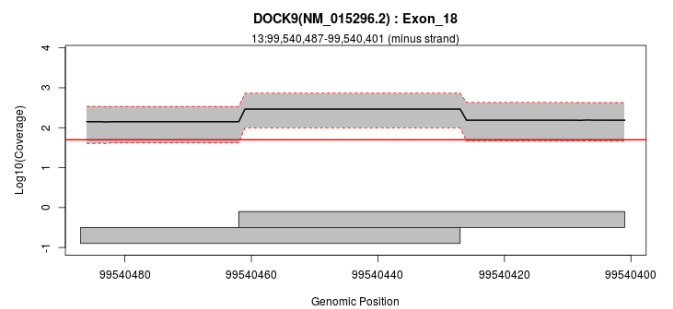
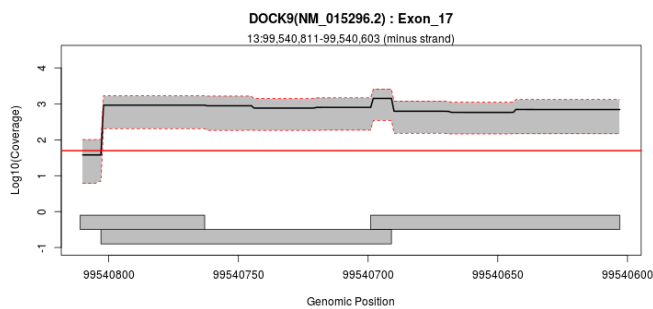
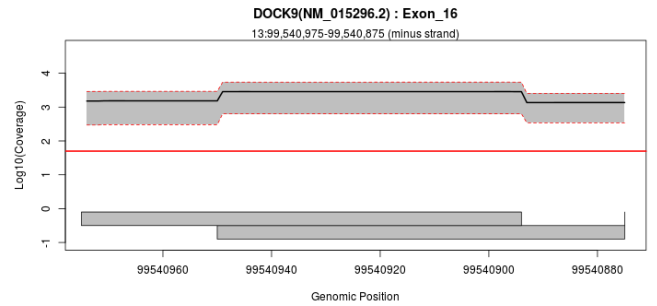
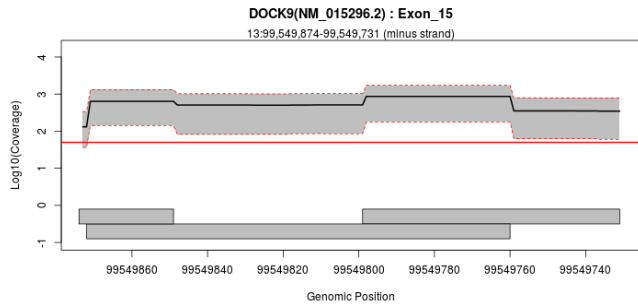
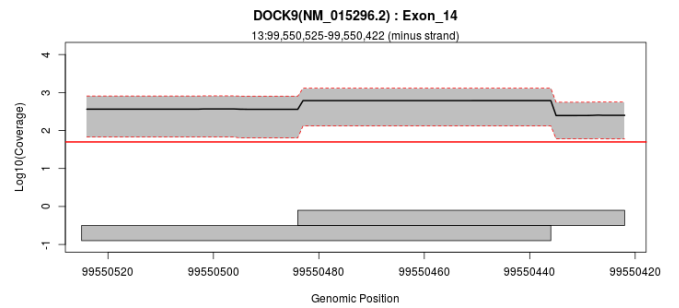
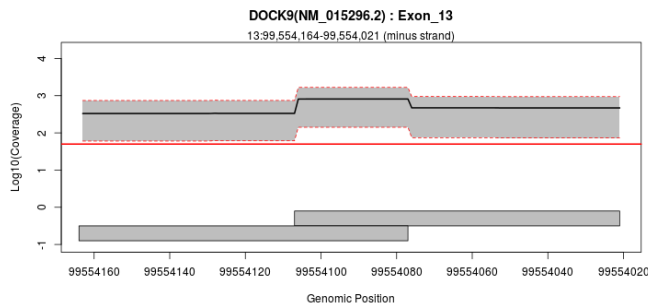
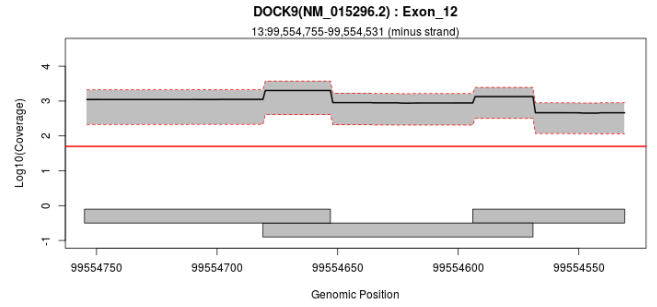
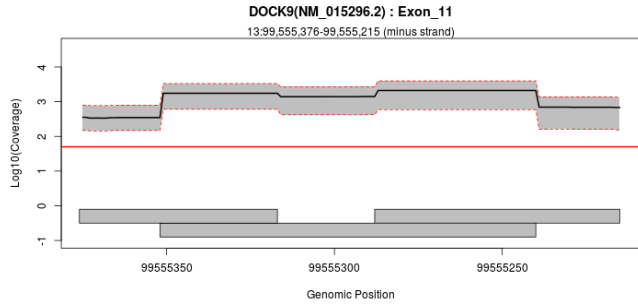
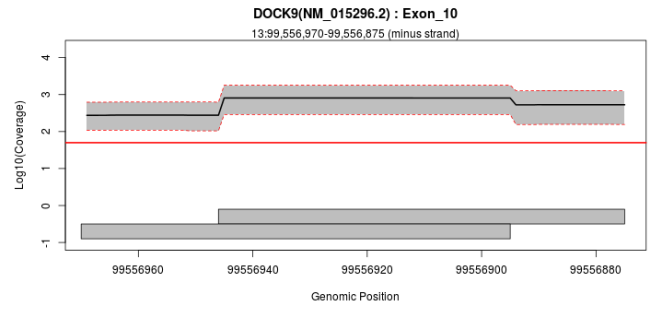
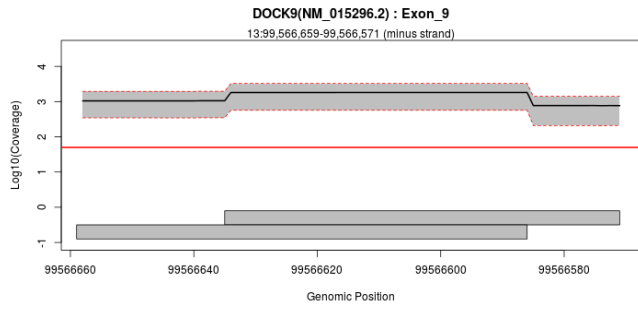


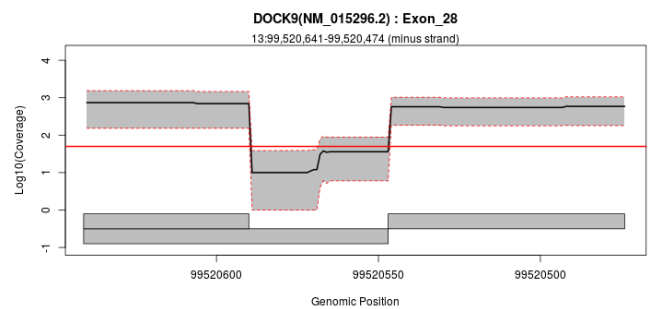
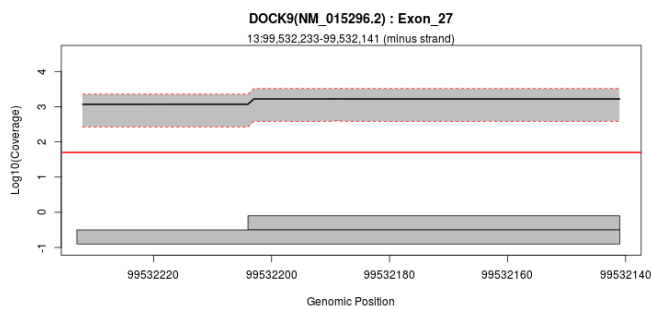
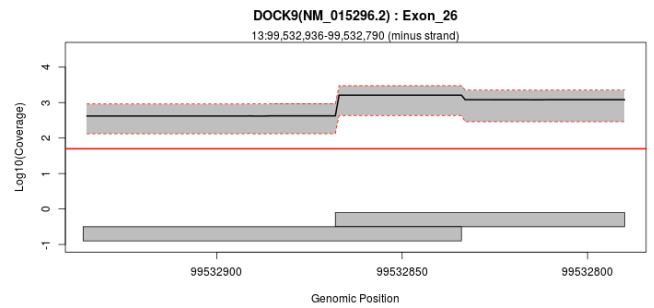
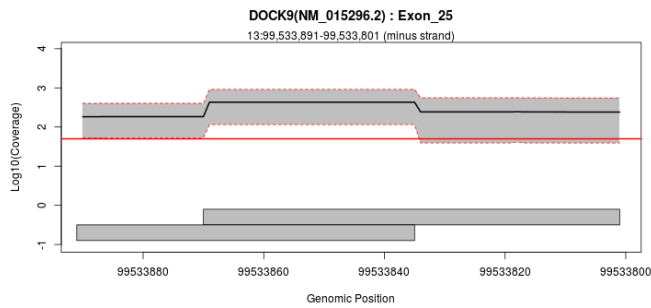
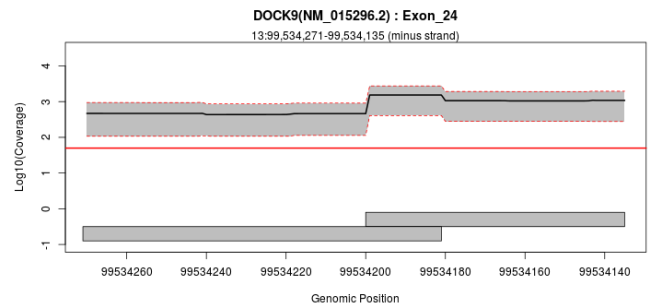
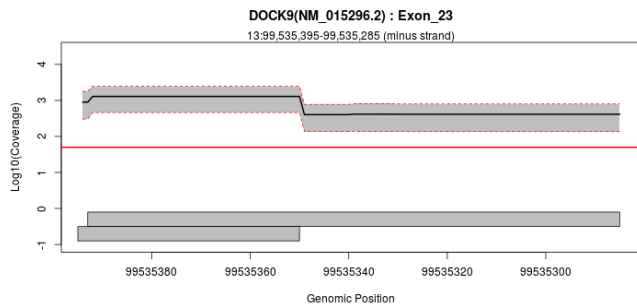
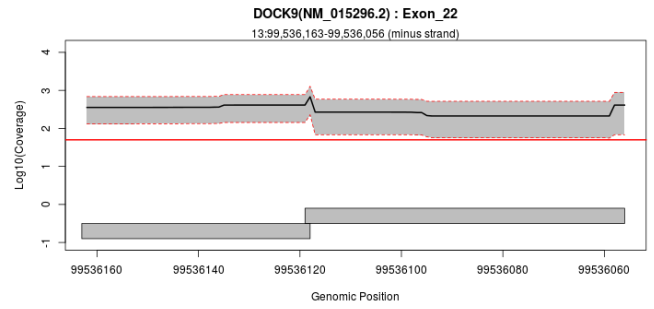
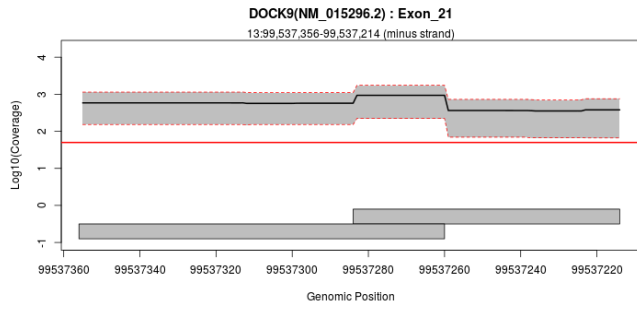
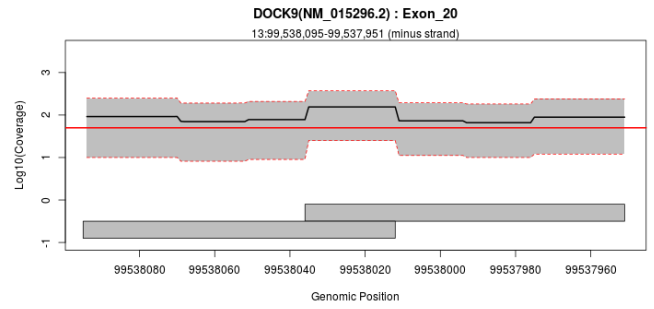
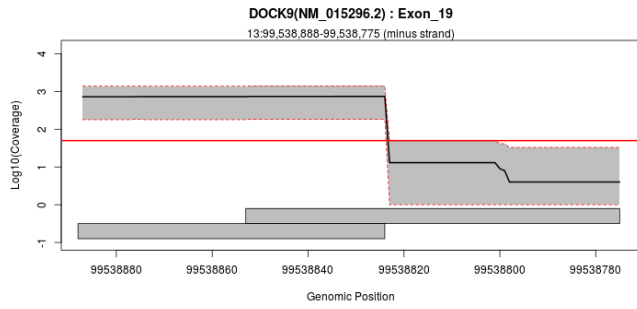


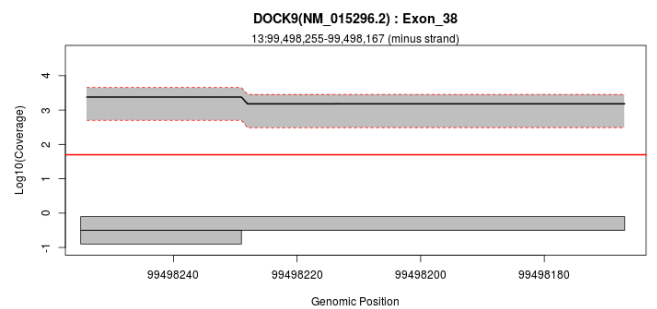
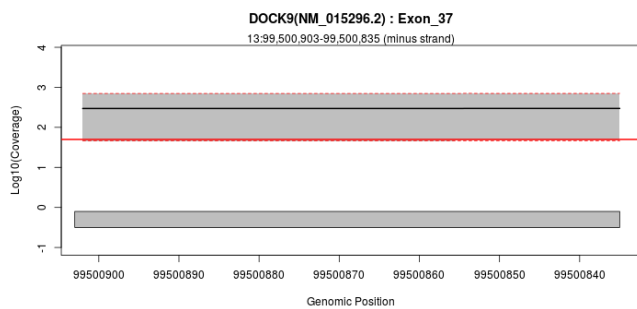
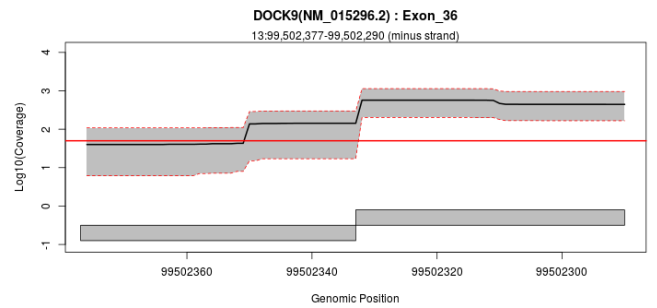
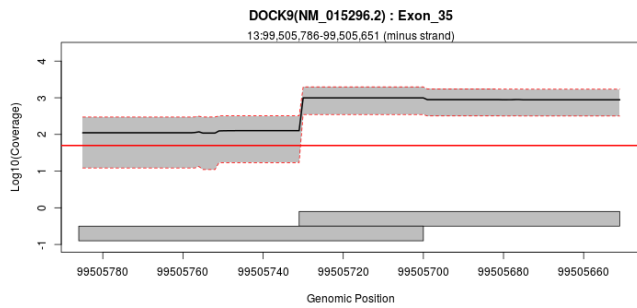
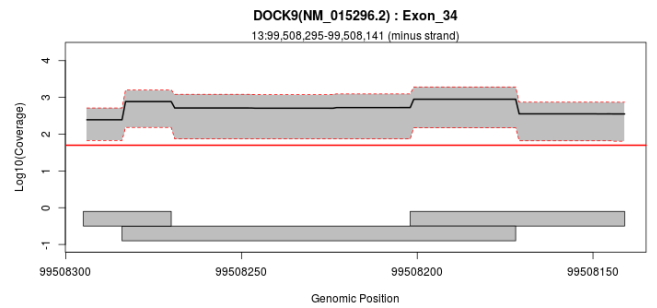
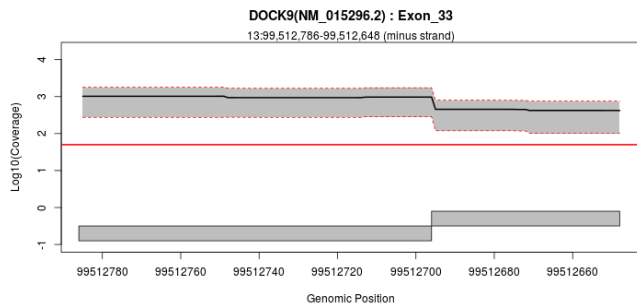
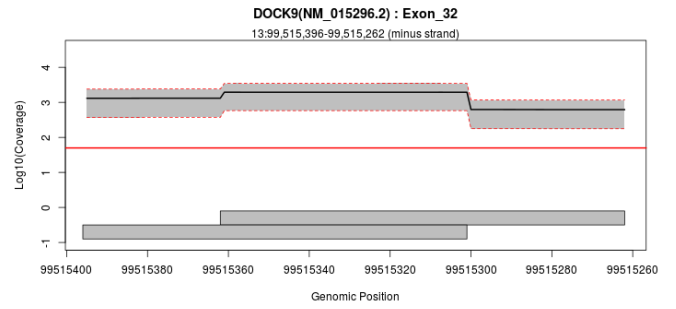
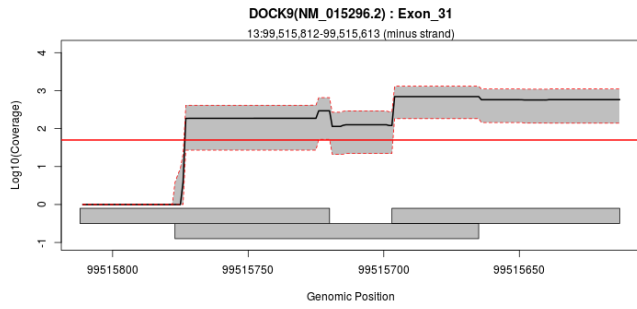
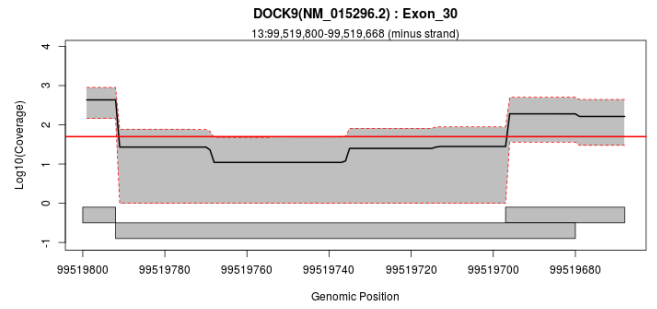
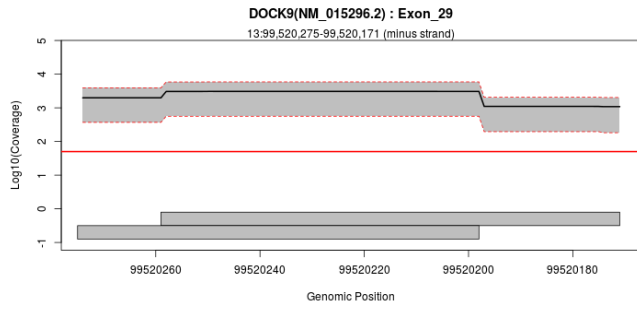


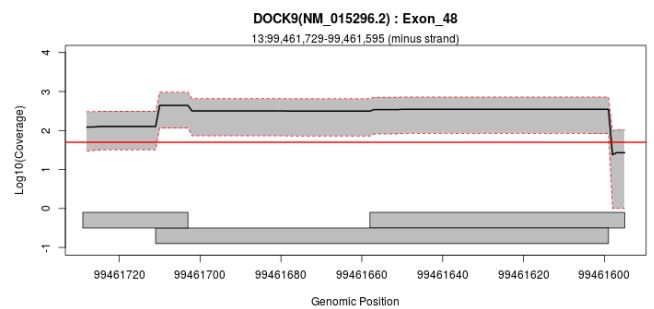
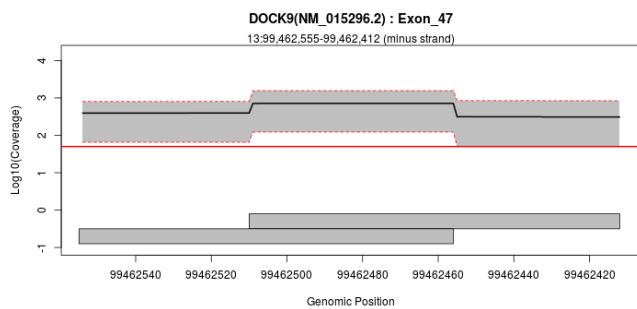
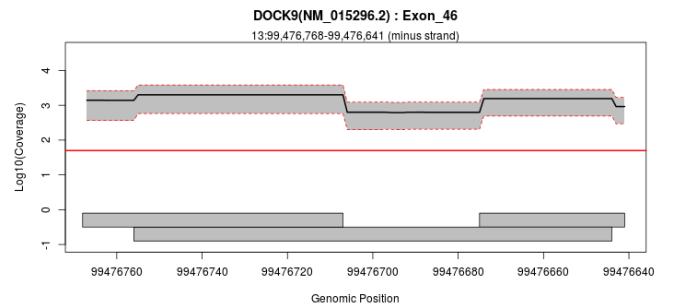
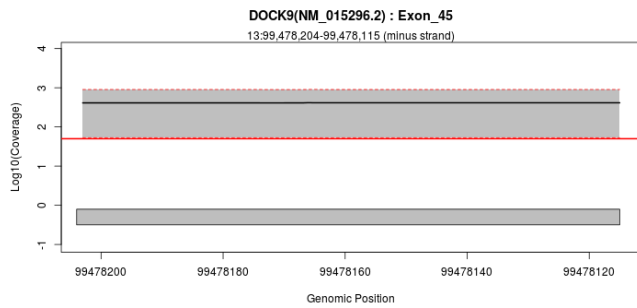
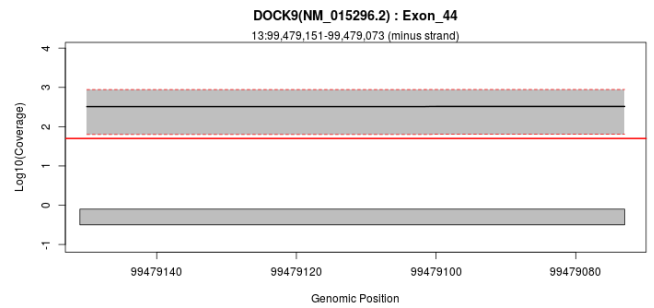
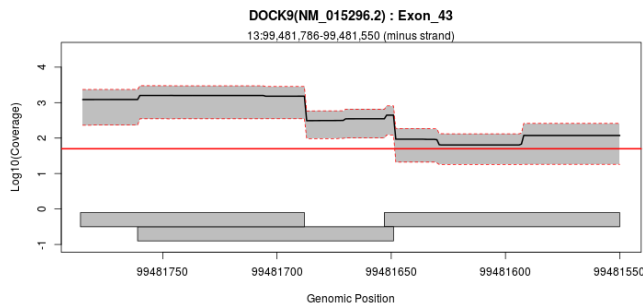
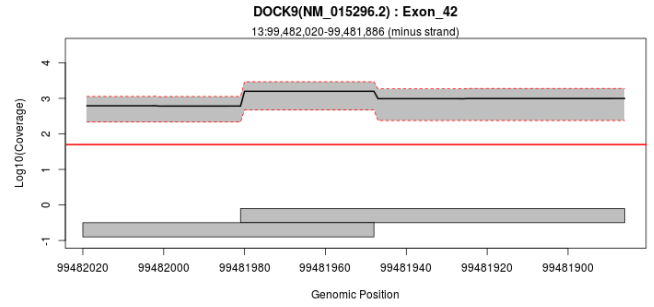
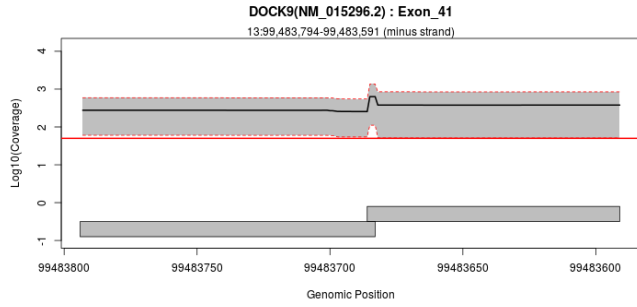
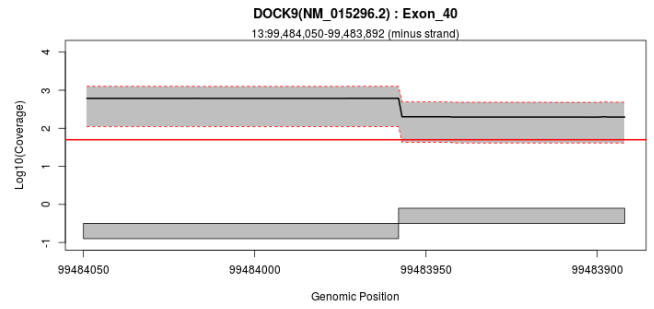
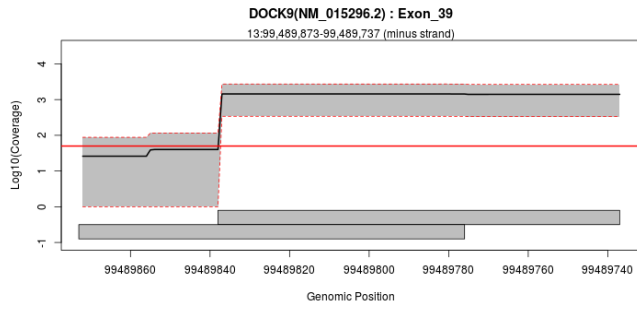
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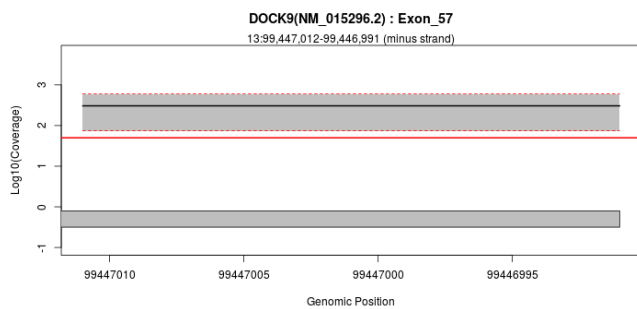
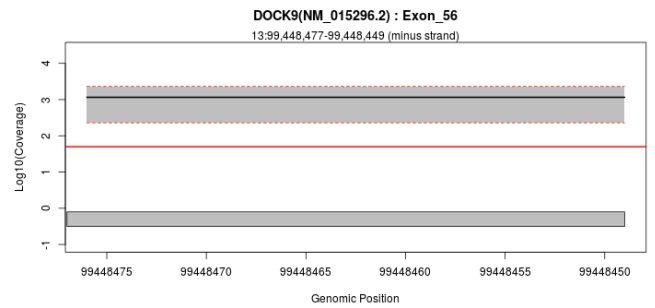
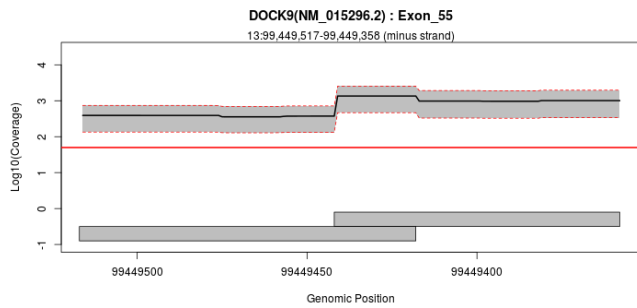
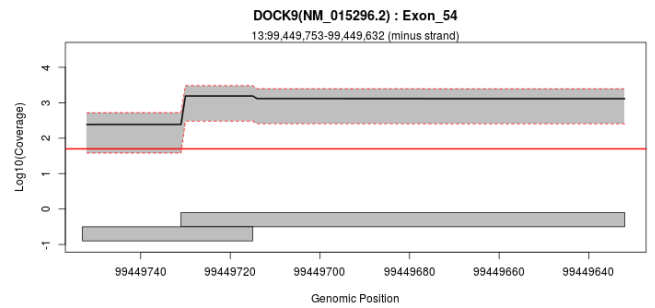
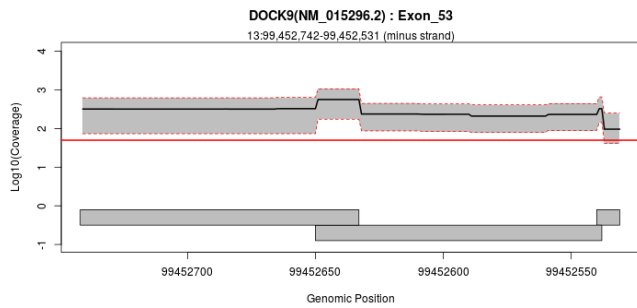
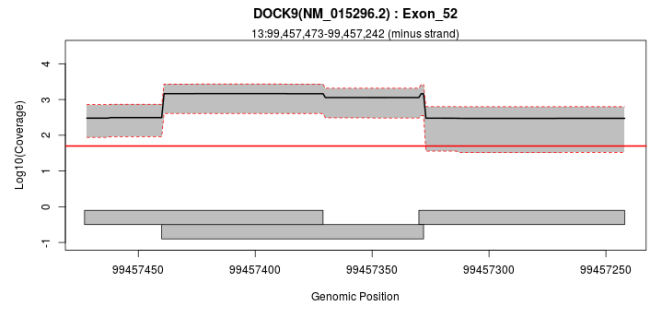
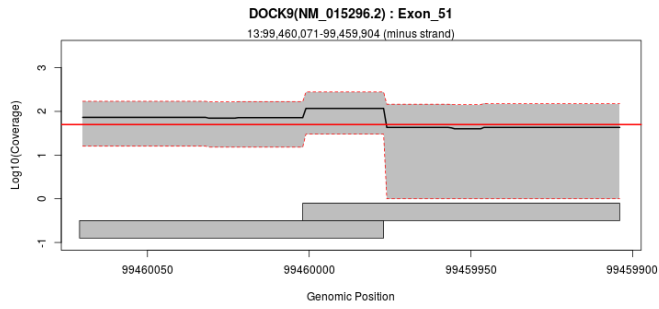
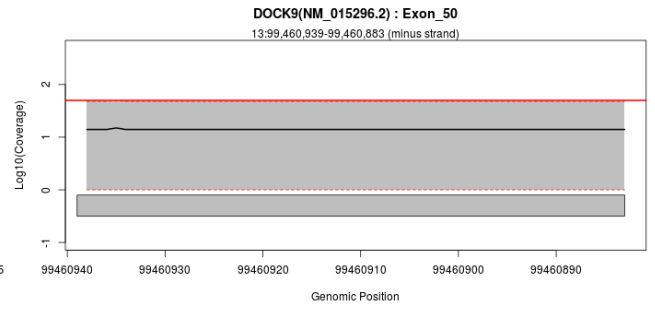
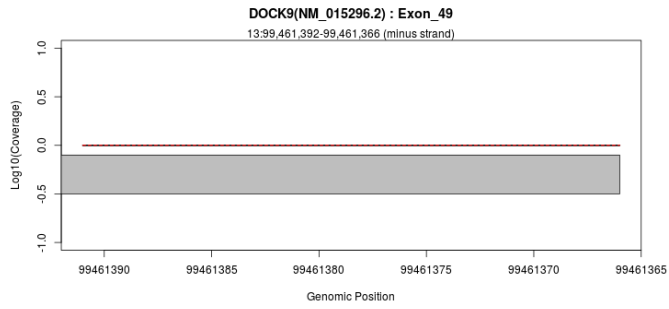




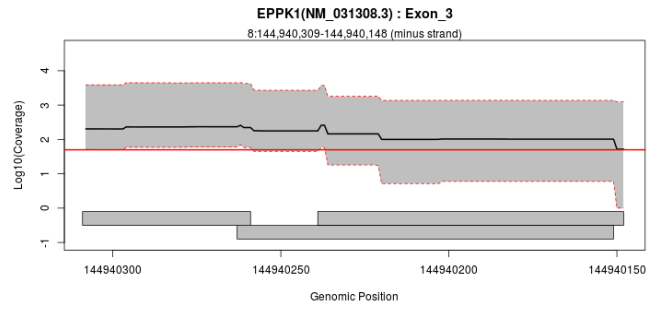
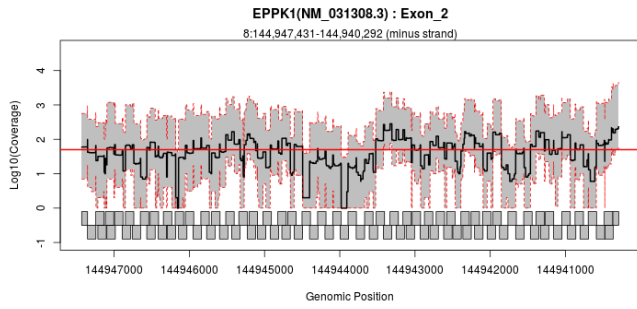




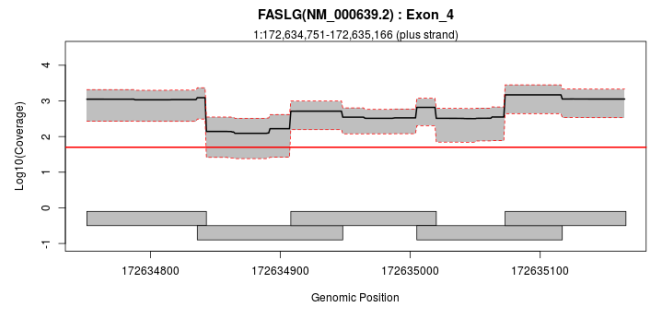
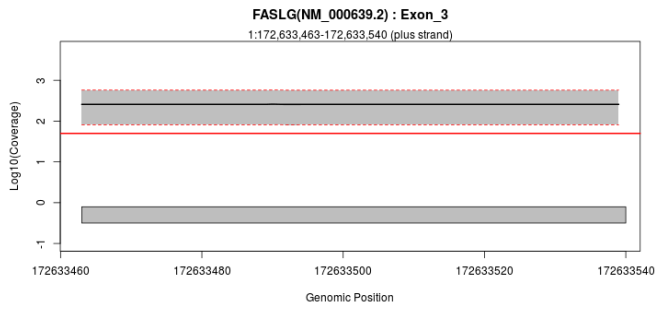
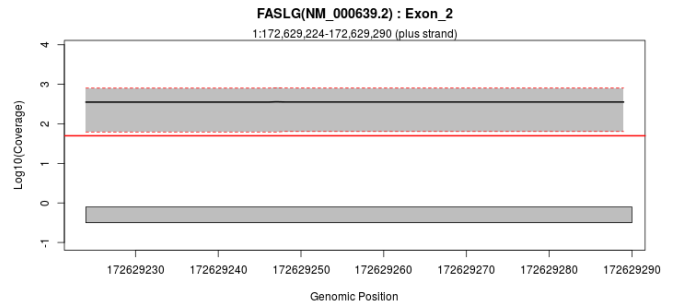
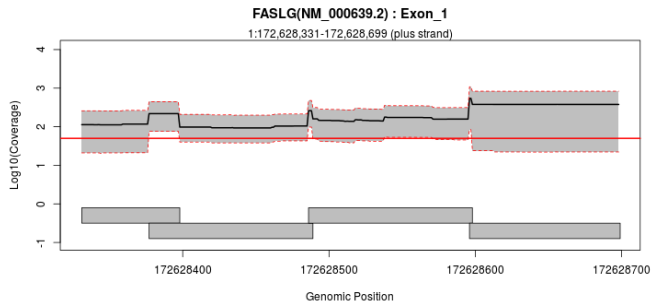




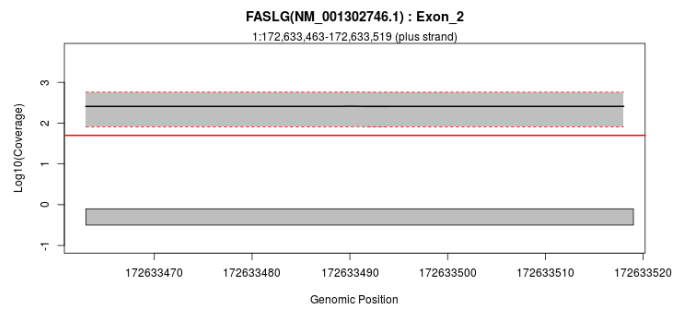
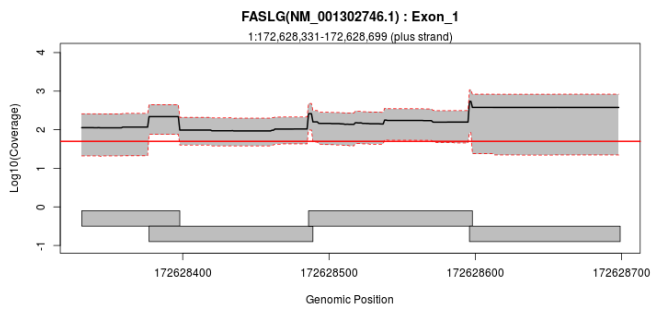
## Exon Overview: EPPK1 : NM\_031308.3



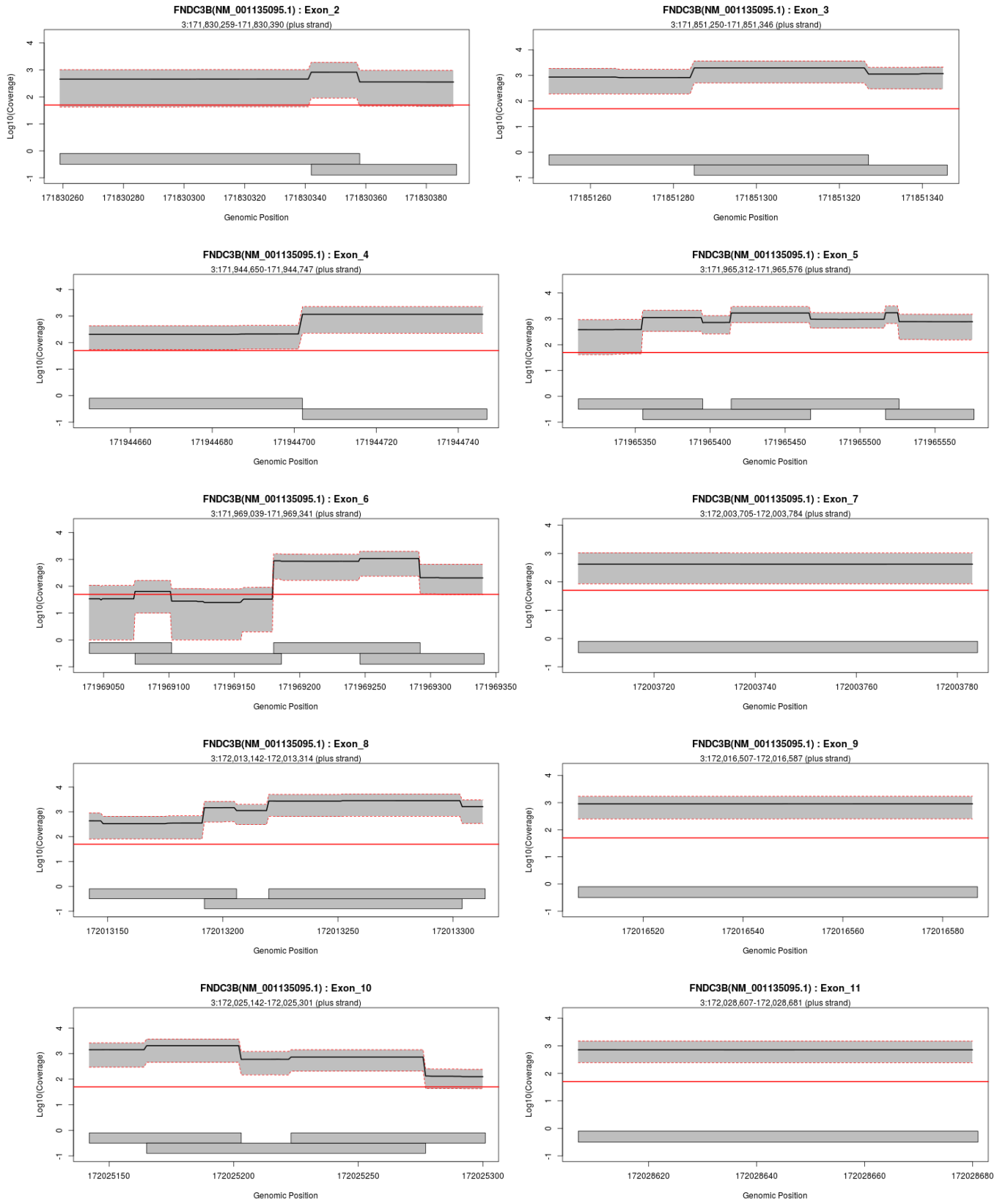
## Exon Overview: FASLG : NM\_000639.2

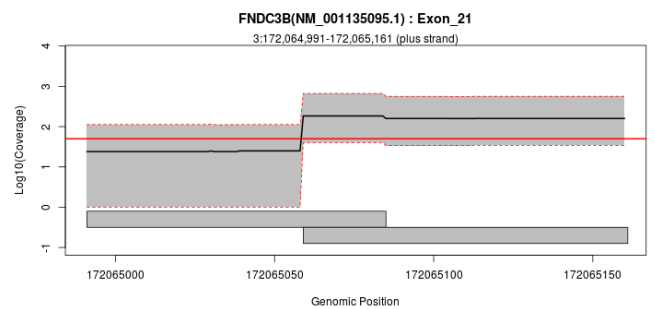
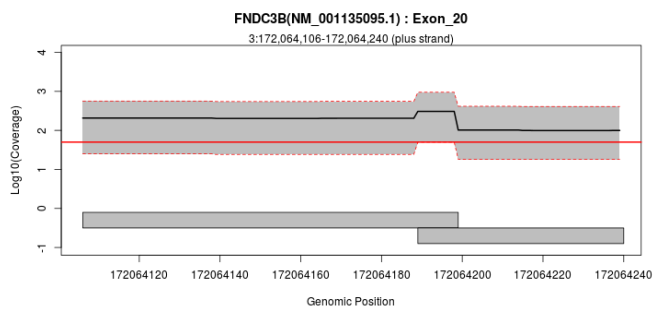
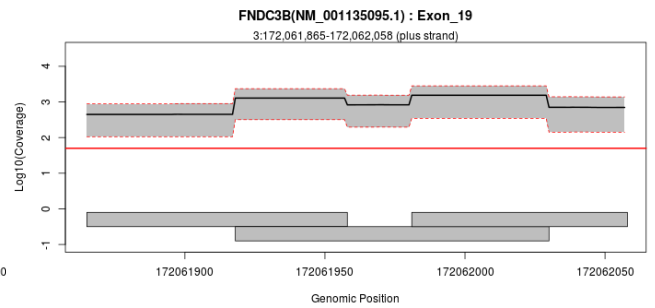
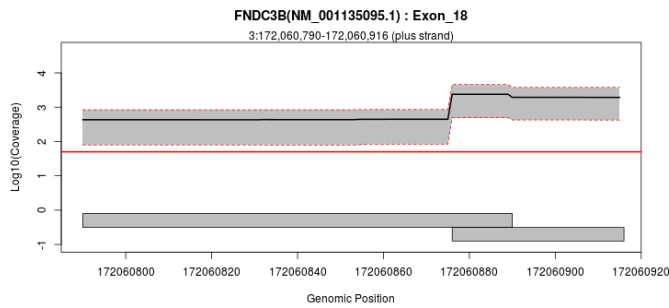
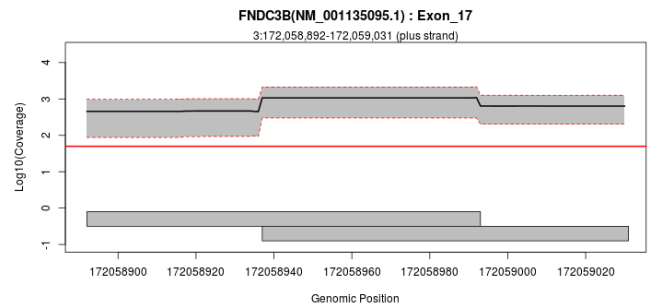
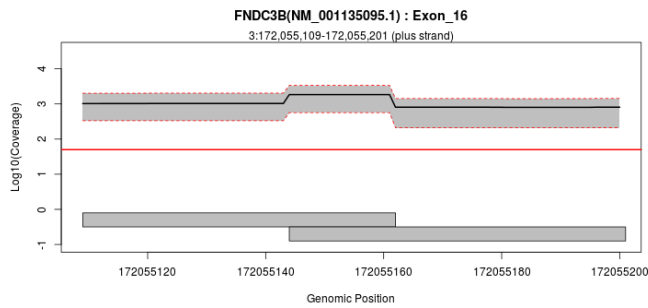
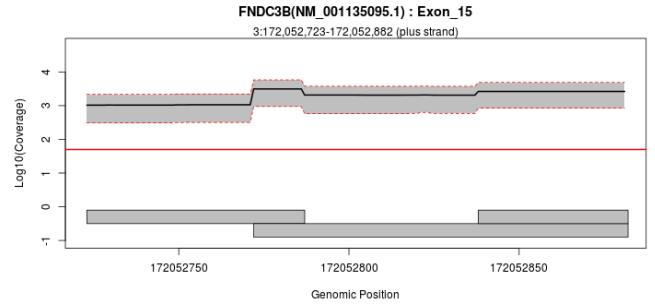
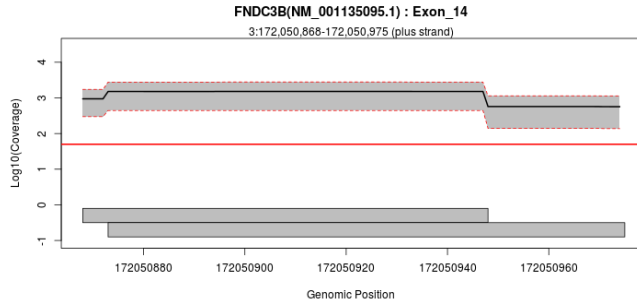
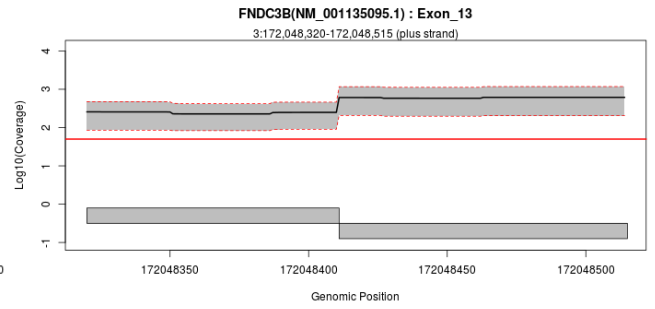
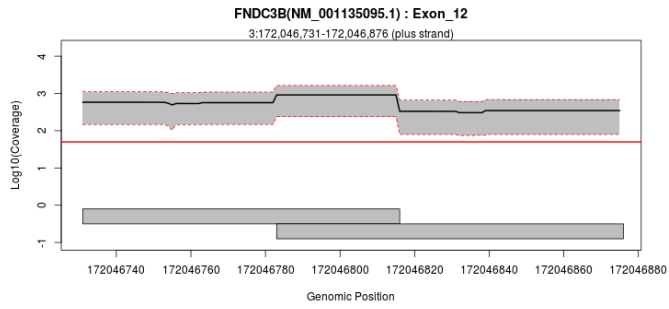


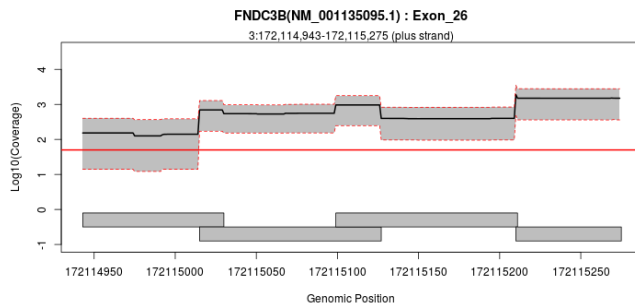
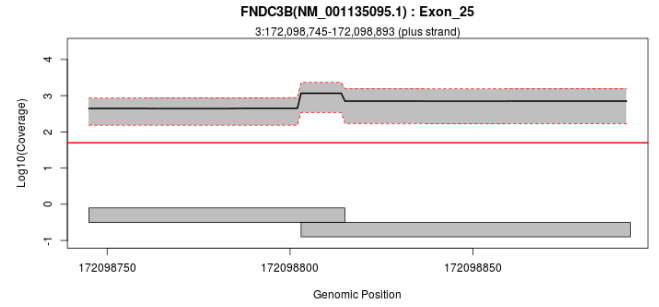
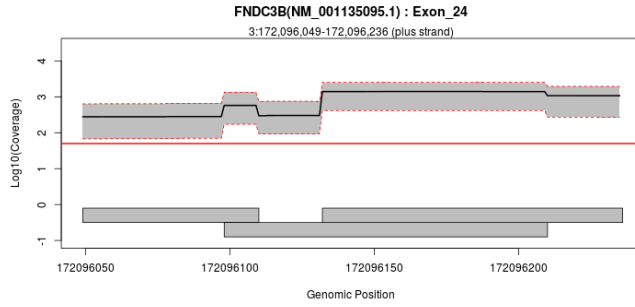
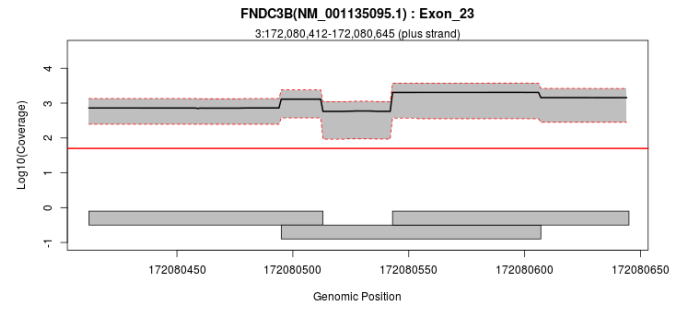
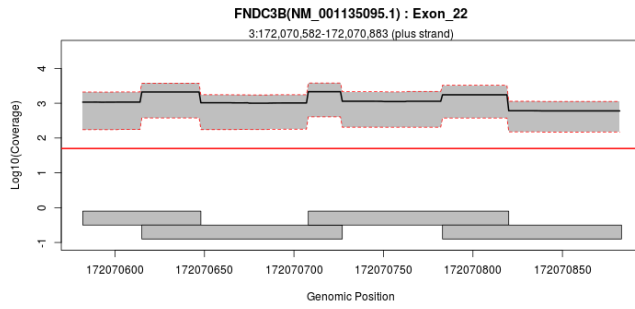
## Exon Overview: FASLG : NM\_001302746.1



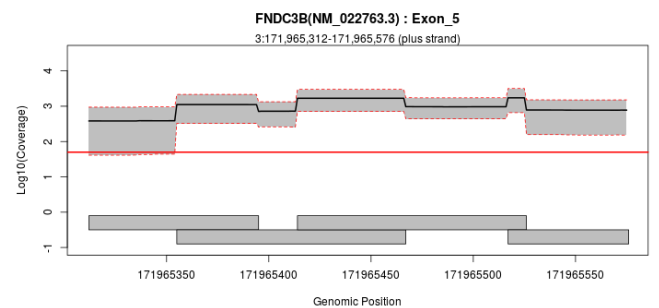
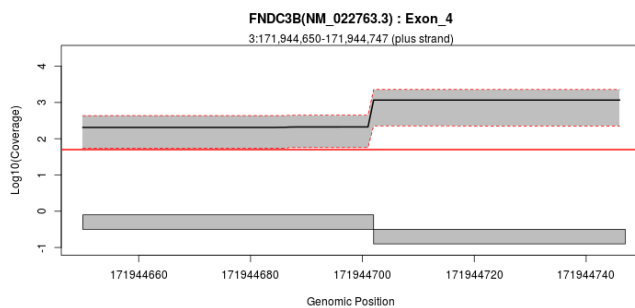
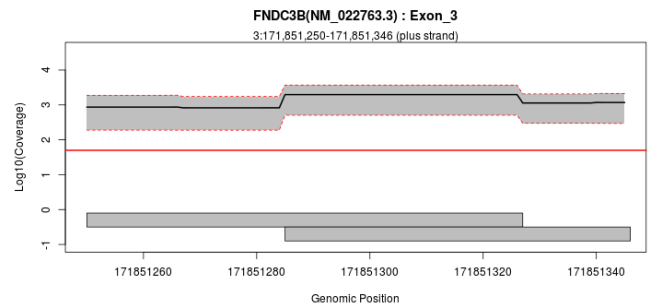
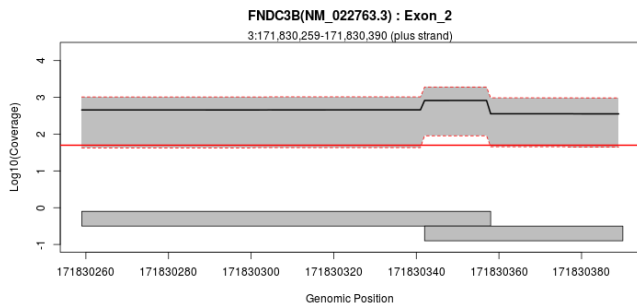
# Exon Overview: FNDC3B : NM\_001135095.1

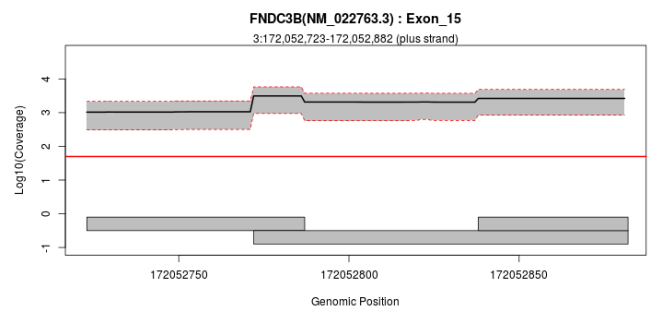
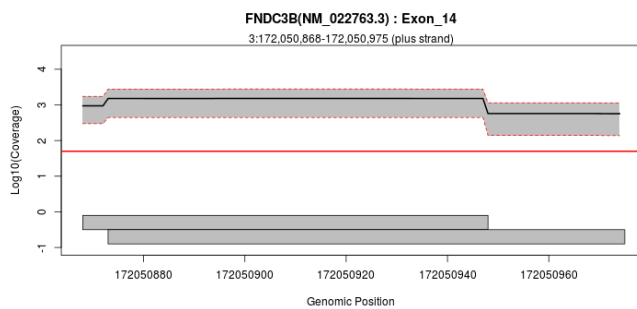
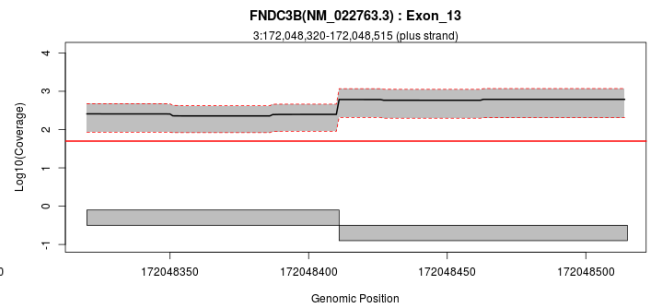
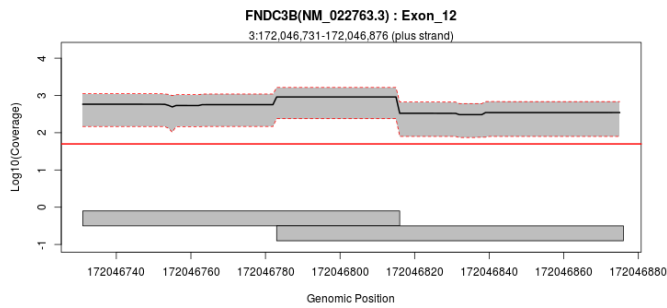
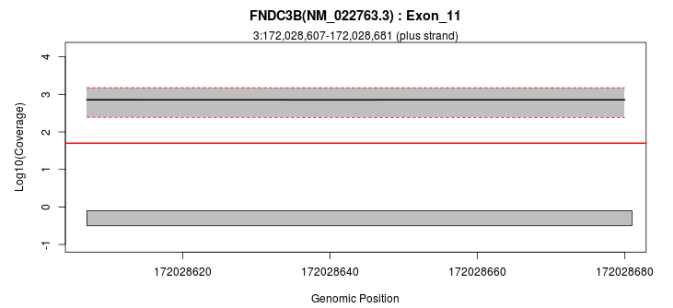
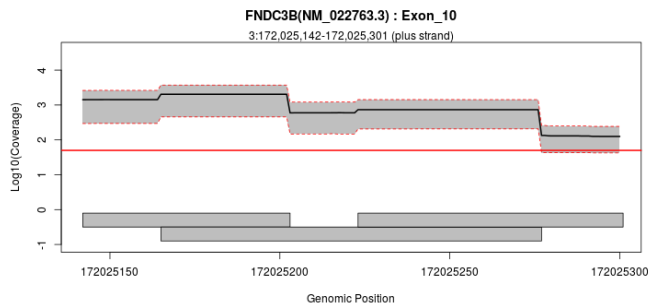
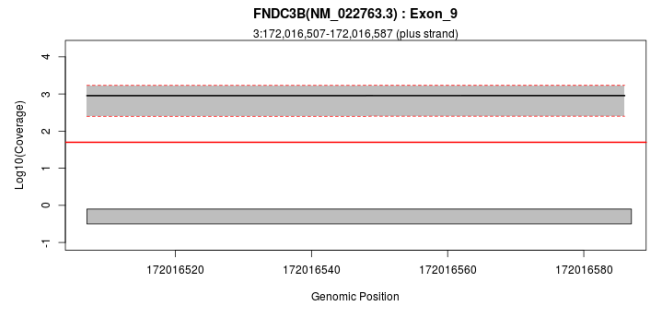
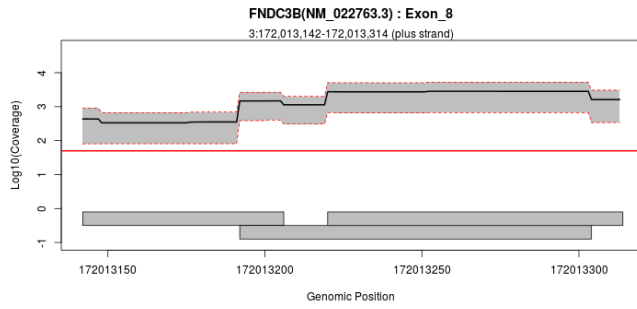
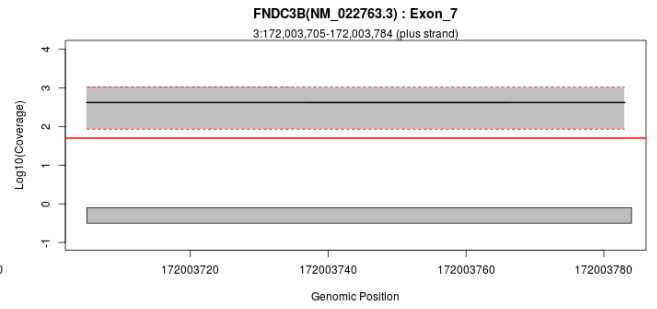
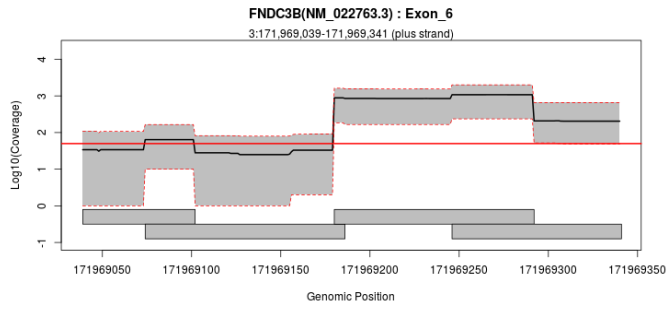


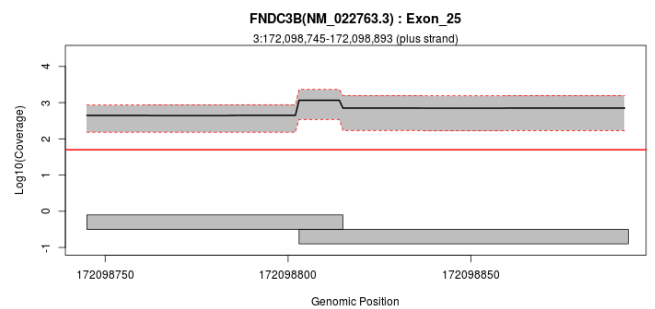
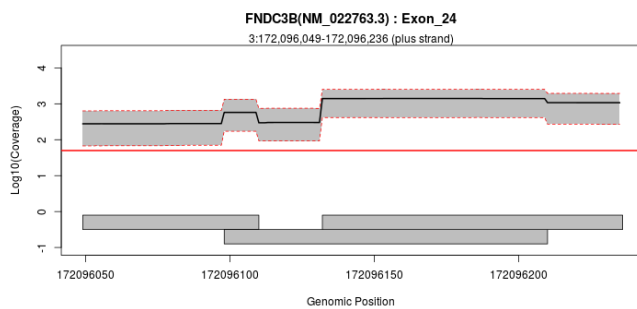
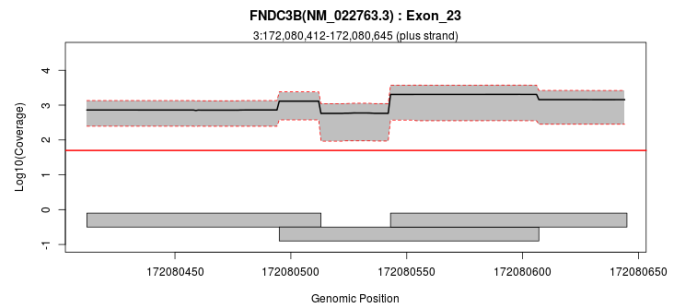
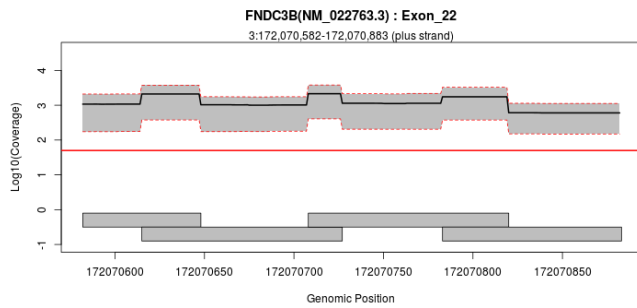
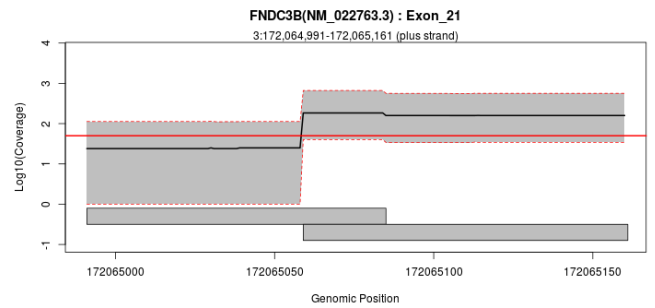
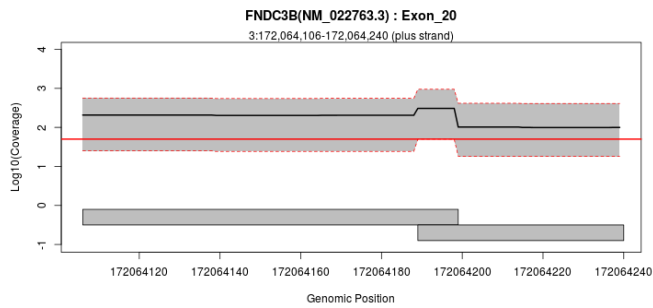
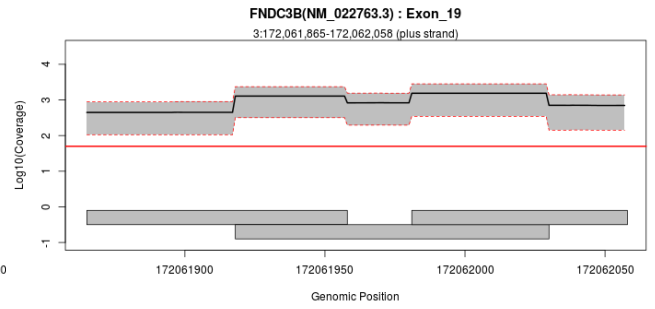
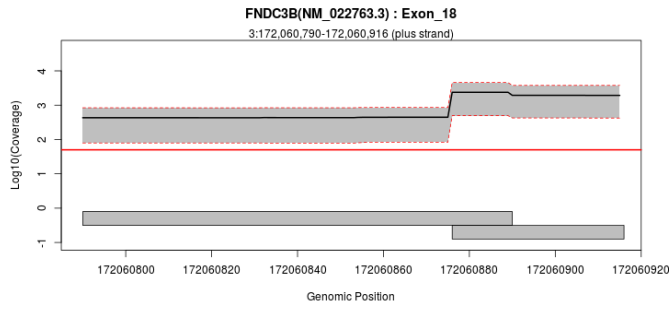
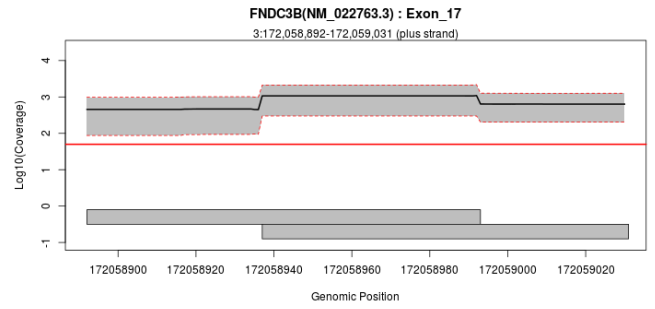
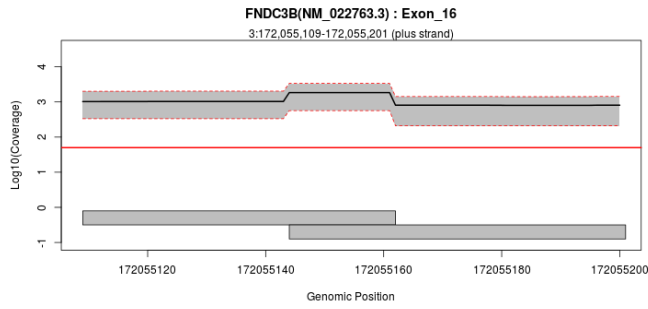


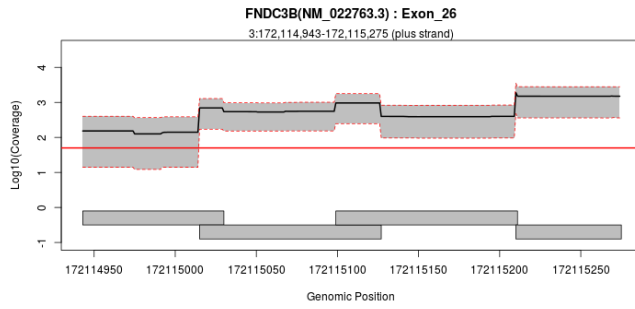


## Exon Overview: FNDC3B : NM\_022763.3

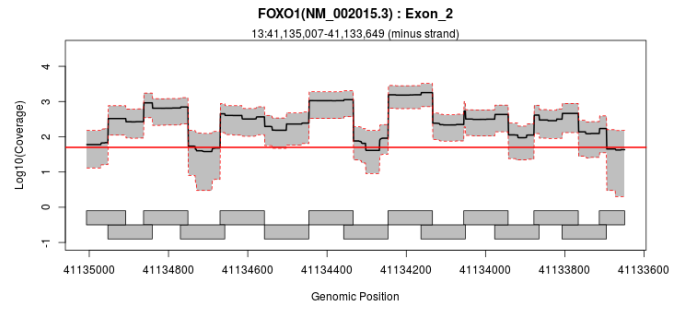
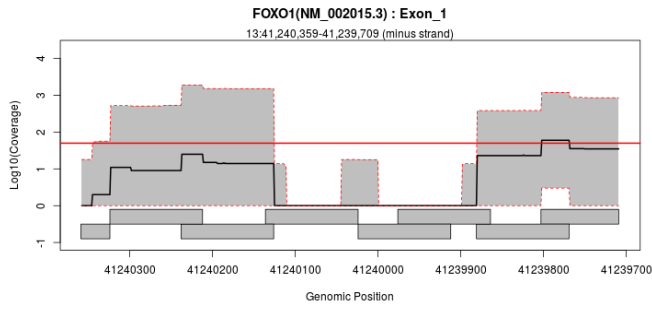




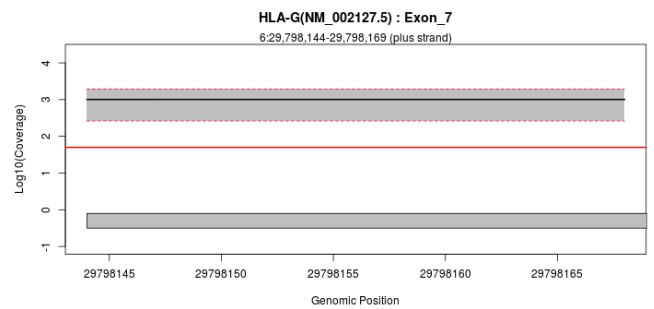
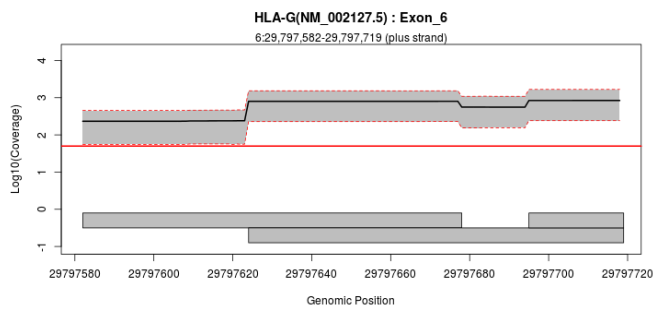
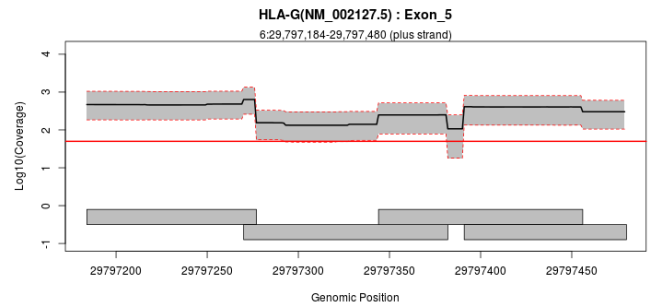
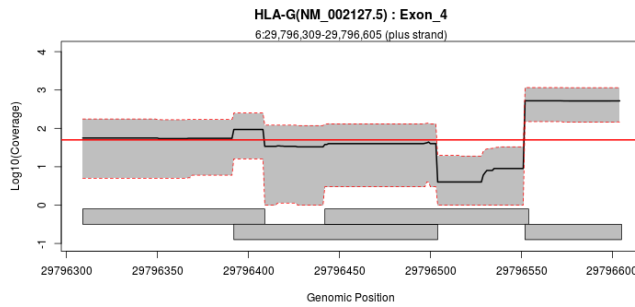
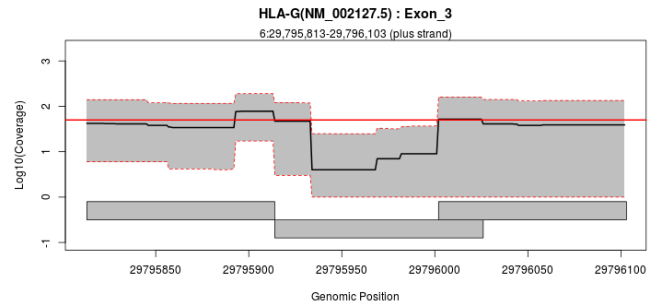
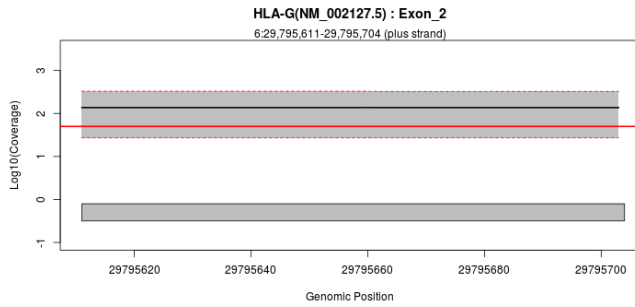




## Exon Overview: FOXO1 : NM\_002015.3

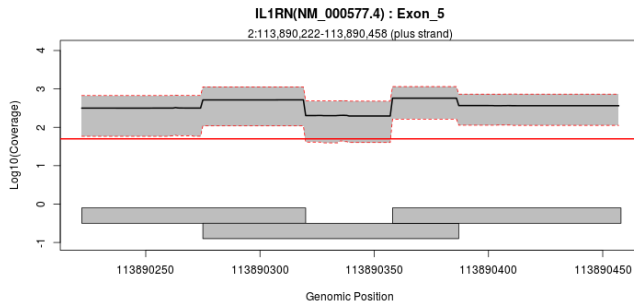
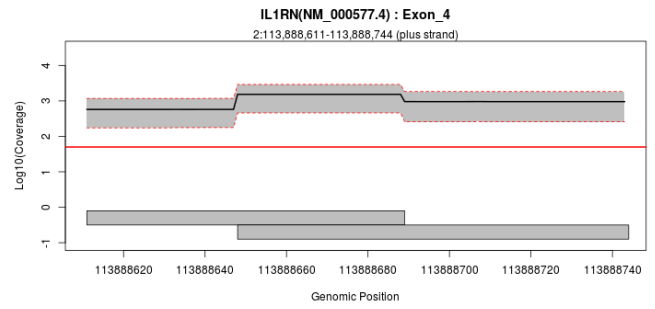
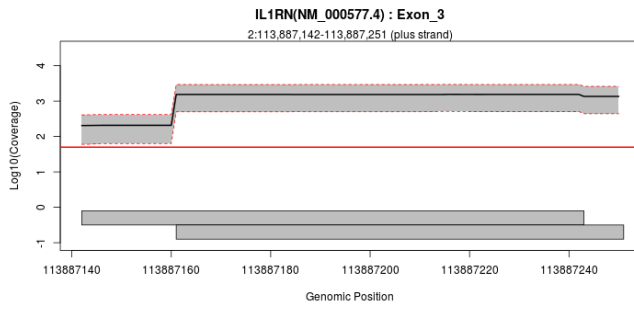
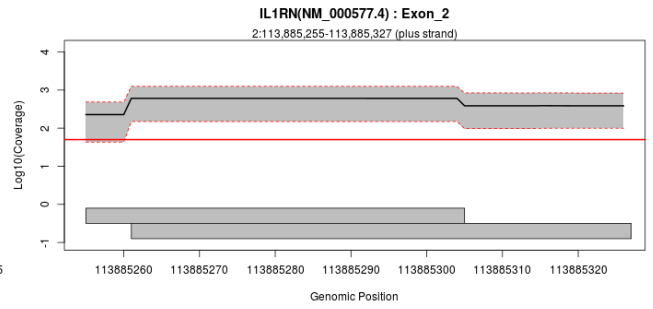
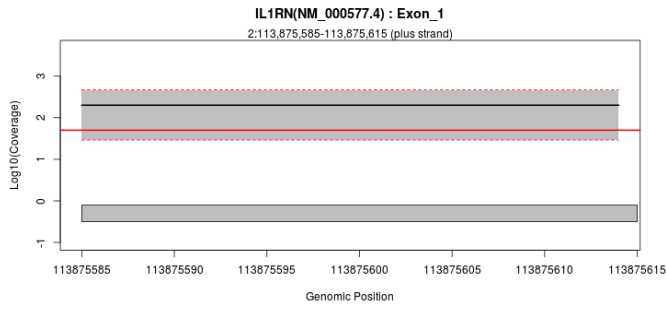


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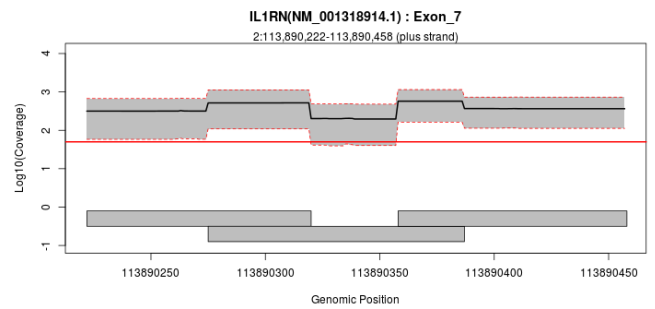
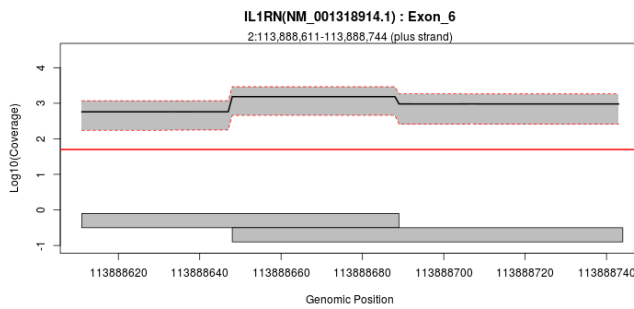
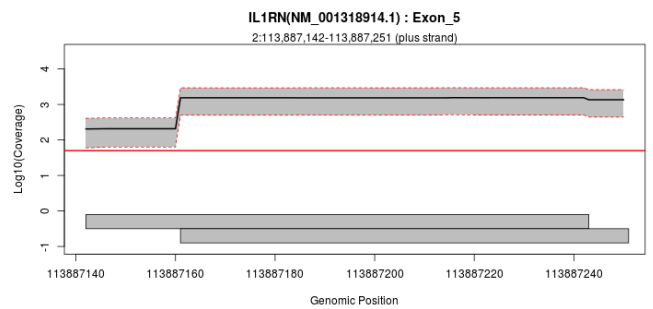
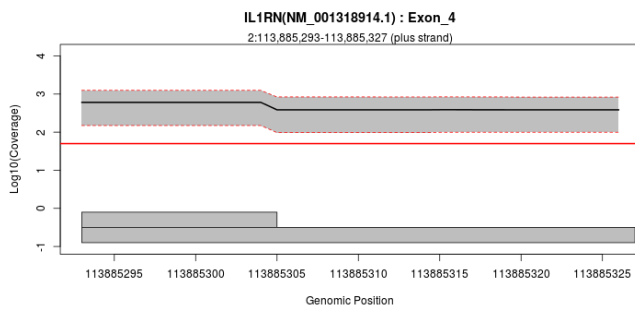




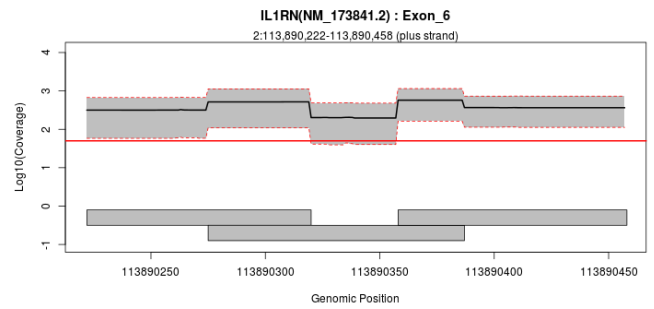
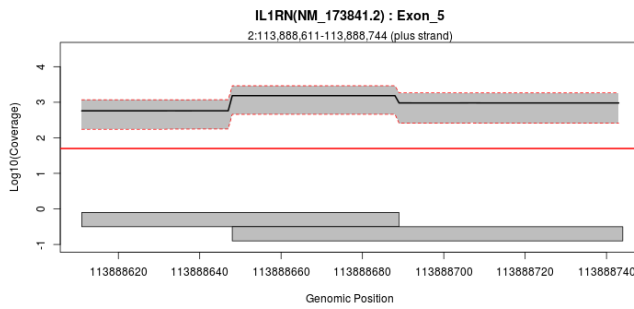
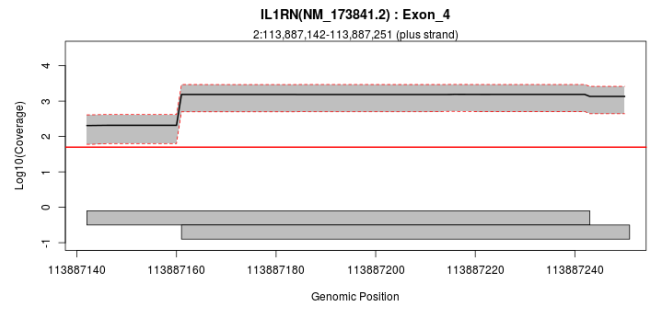
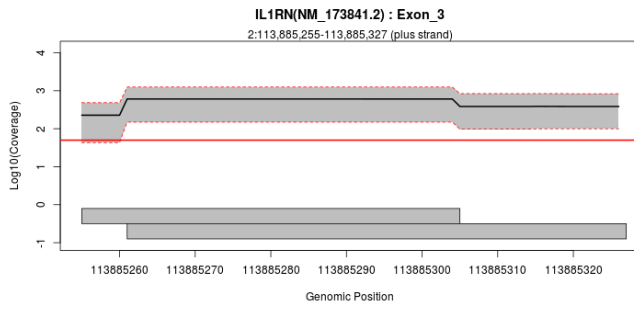
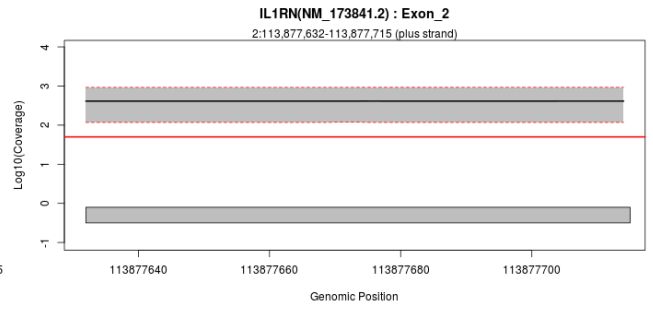
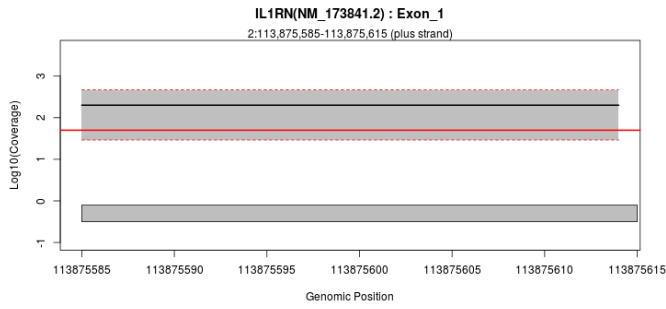
## Exon Overview: IL1RN : NM\_000577.4



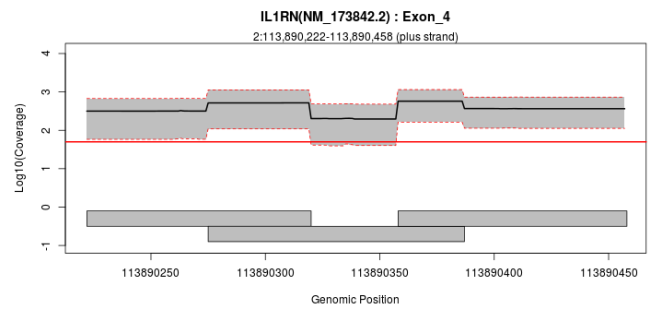
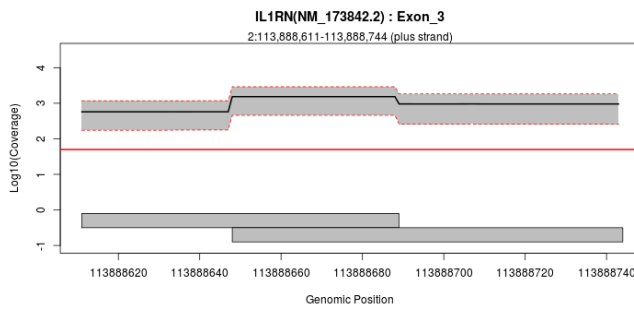
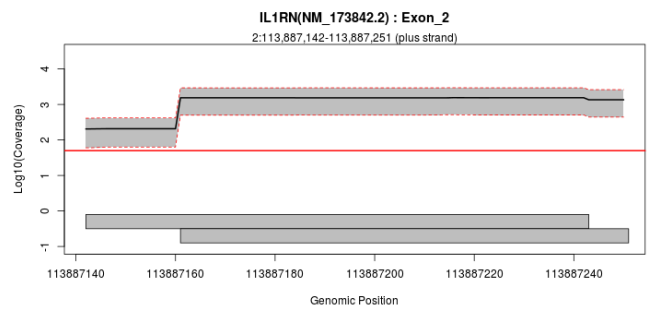
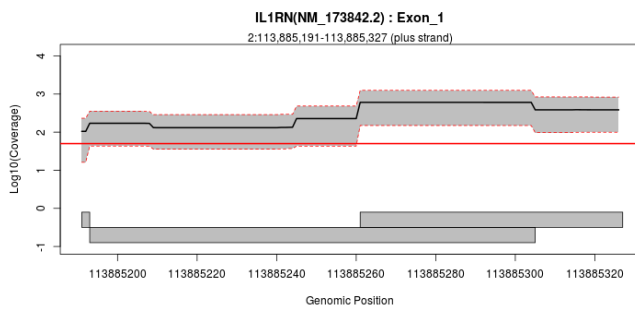
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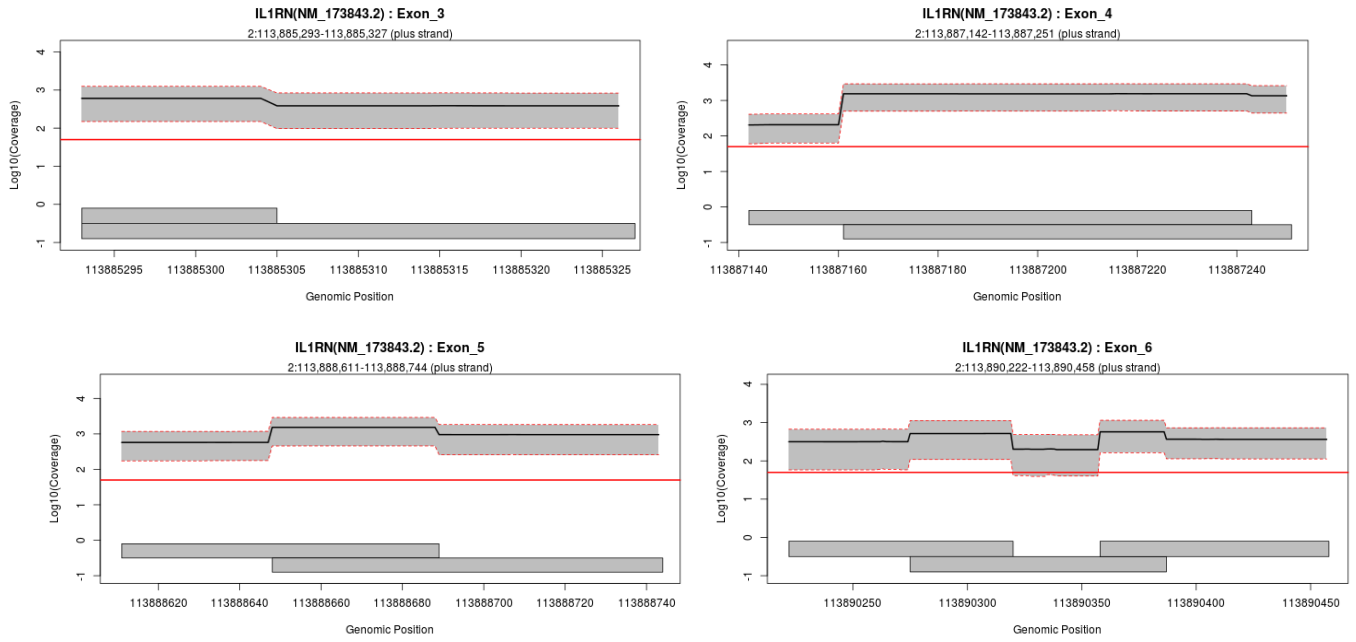
## Exon Overview: IL1RN : NM\_173841.2



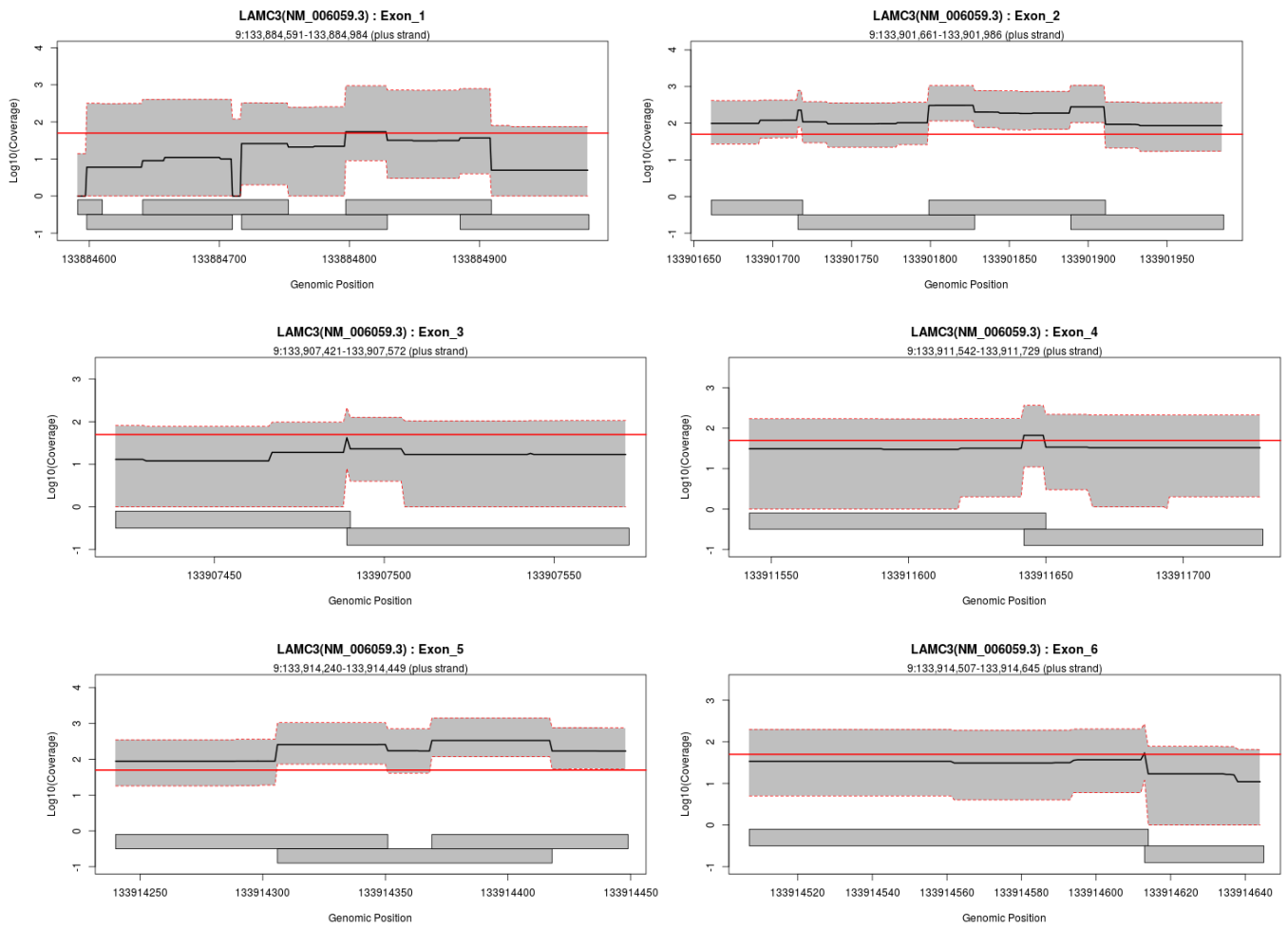
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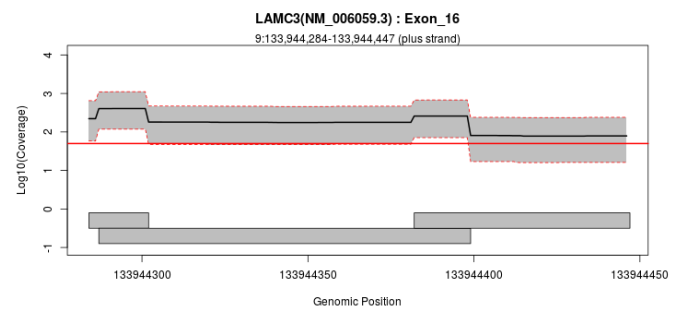
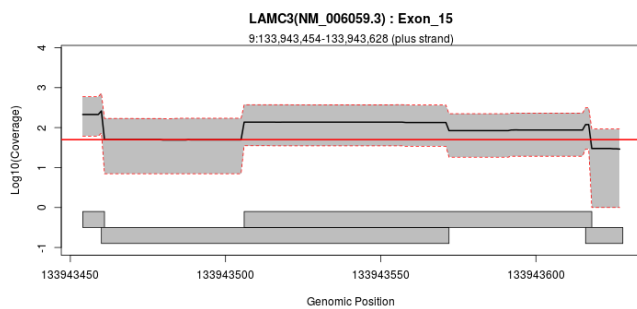
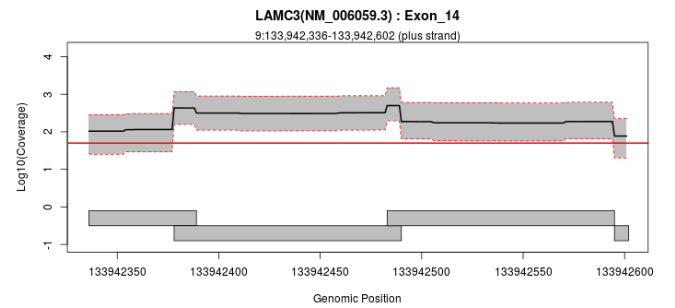
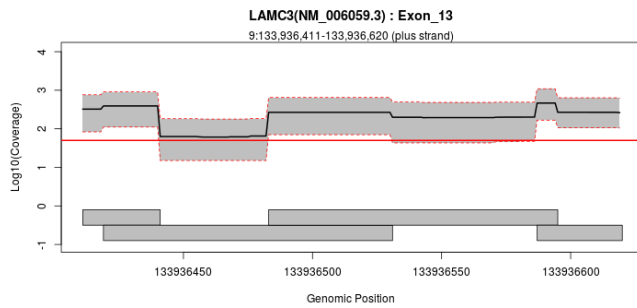
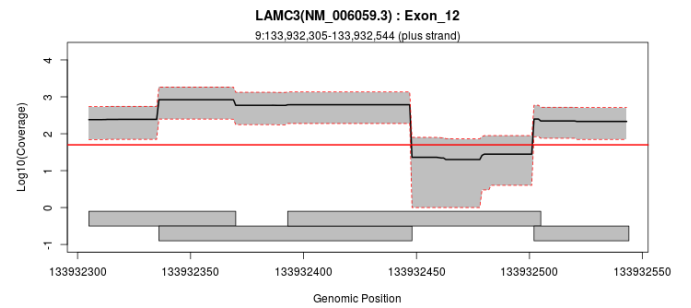
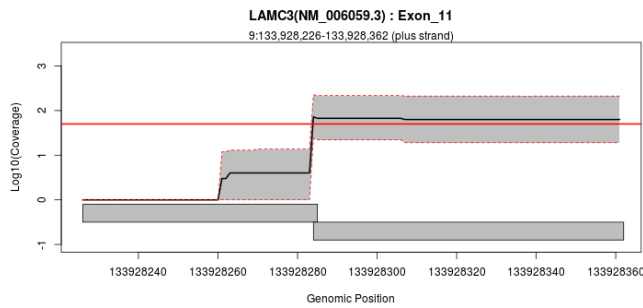
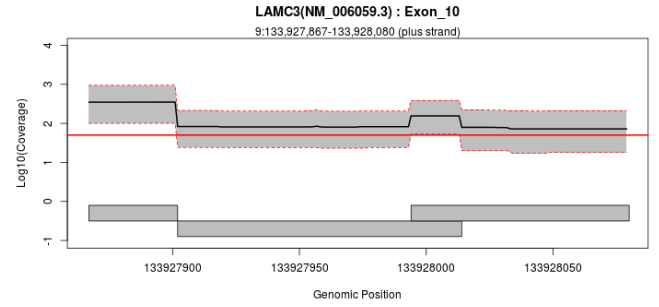
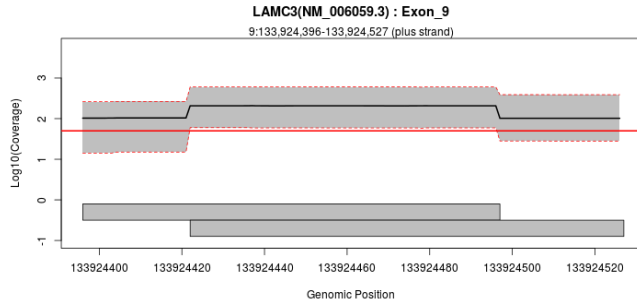
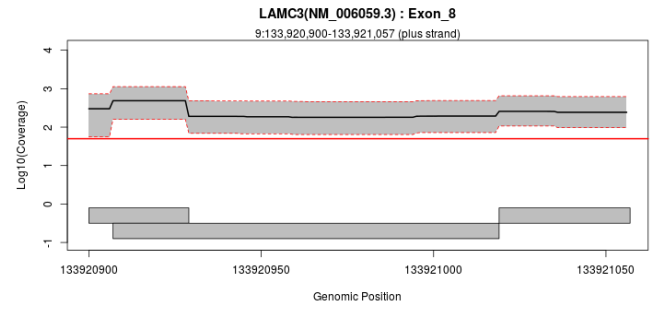
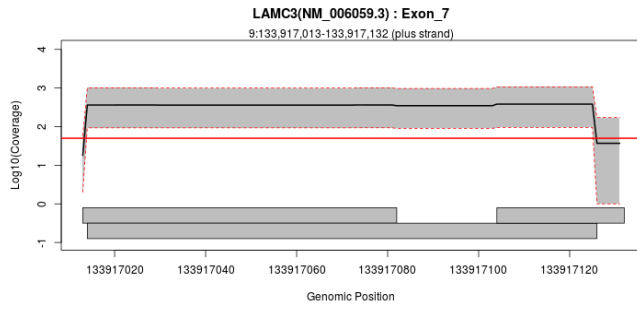


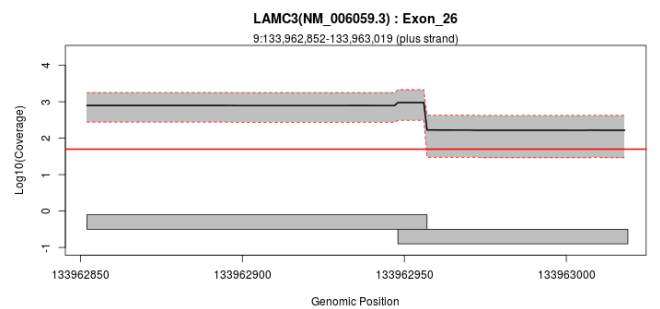
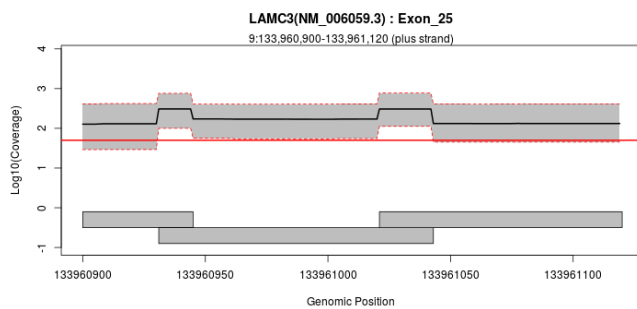
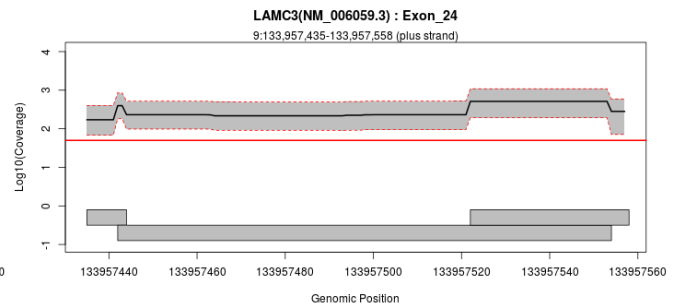
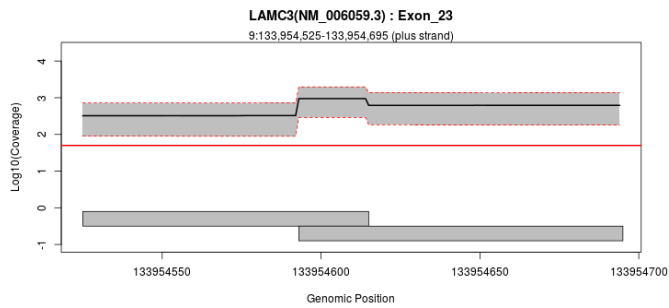
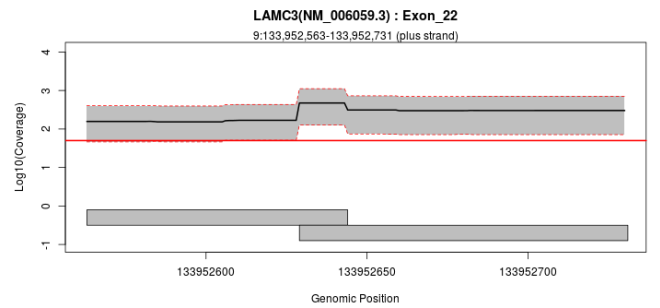
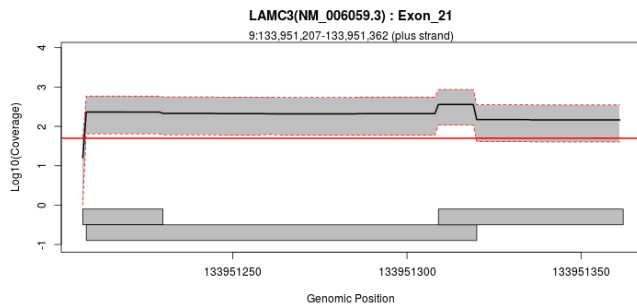
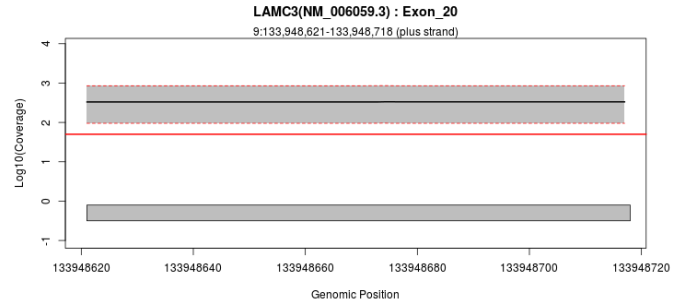
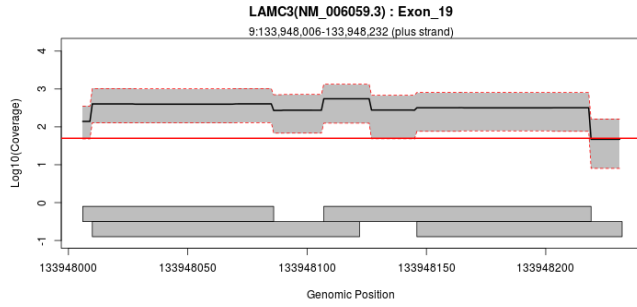
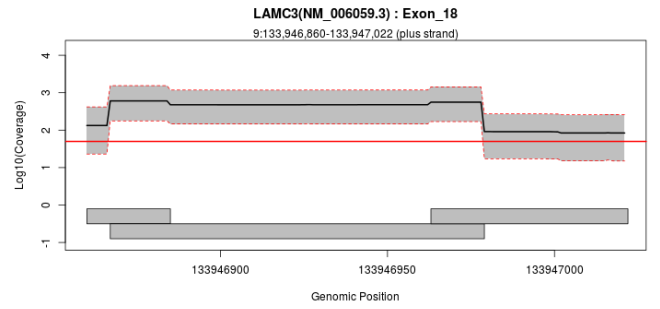
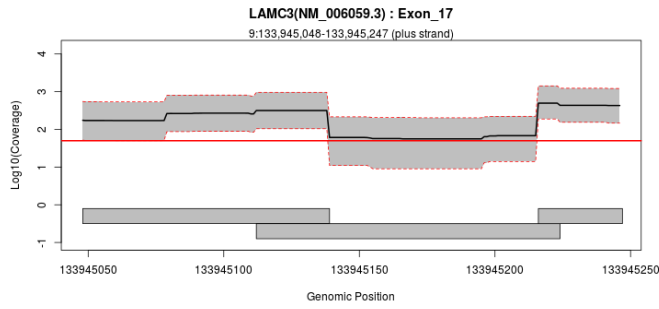
## Exon Overview: IL1RN : NM\_173843.2

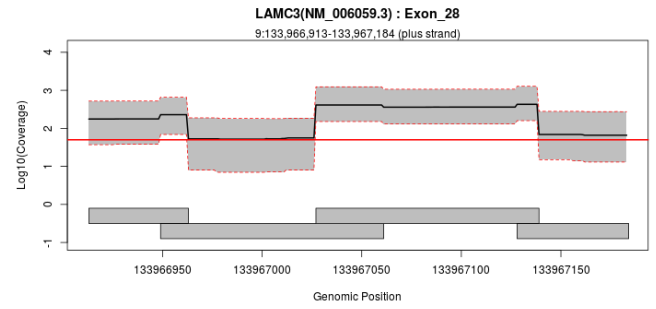
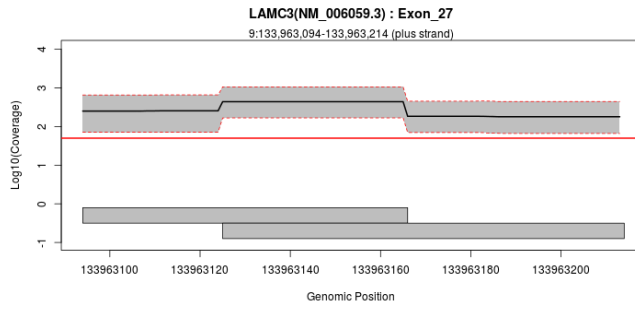


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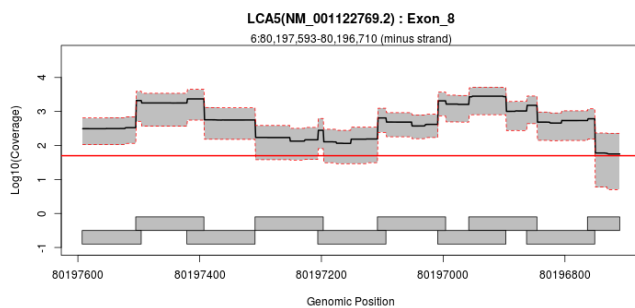
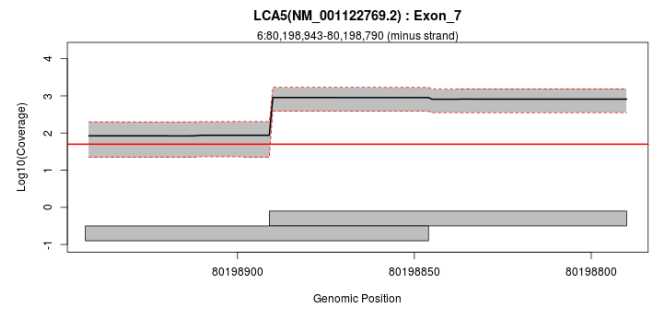
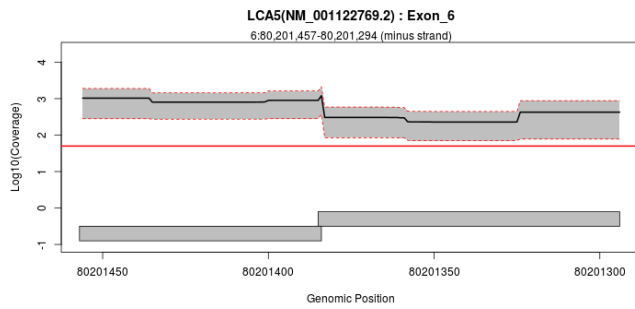
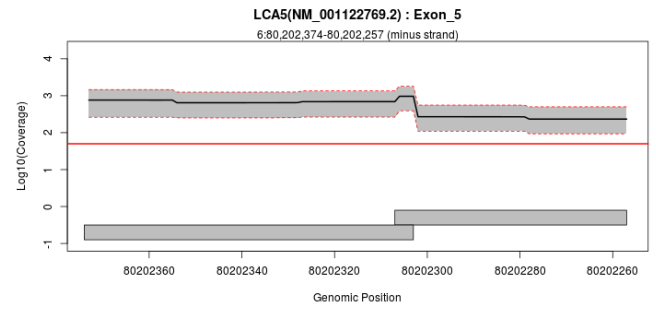
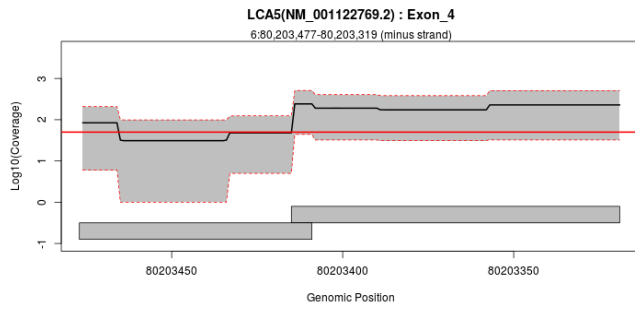
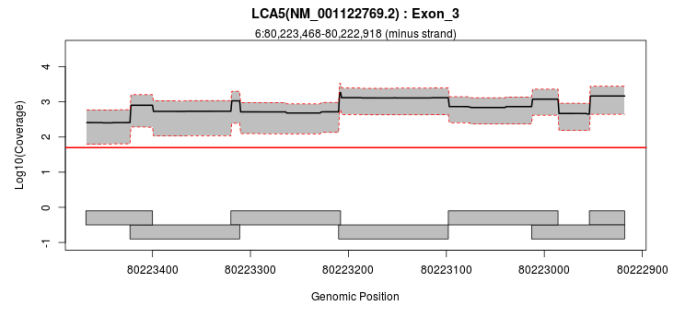
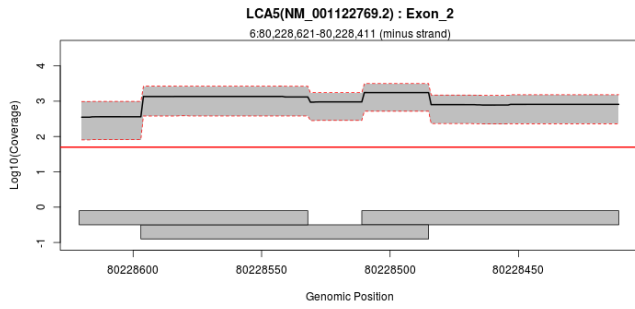




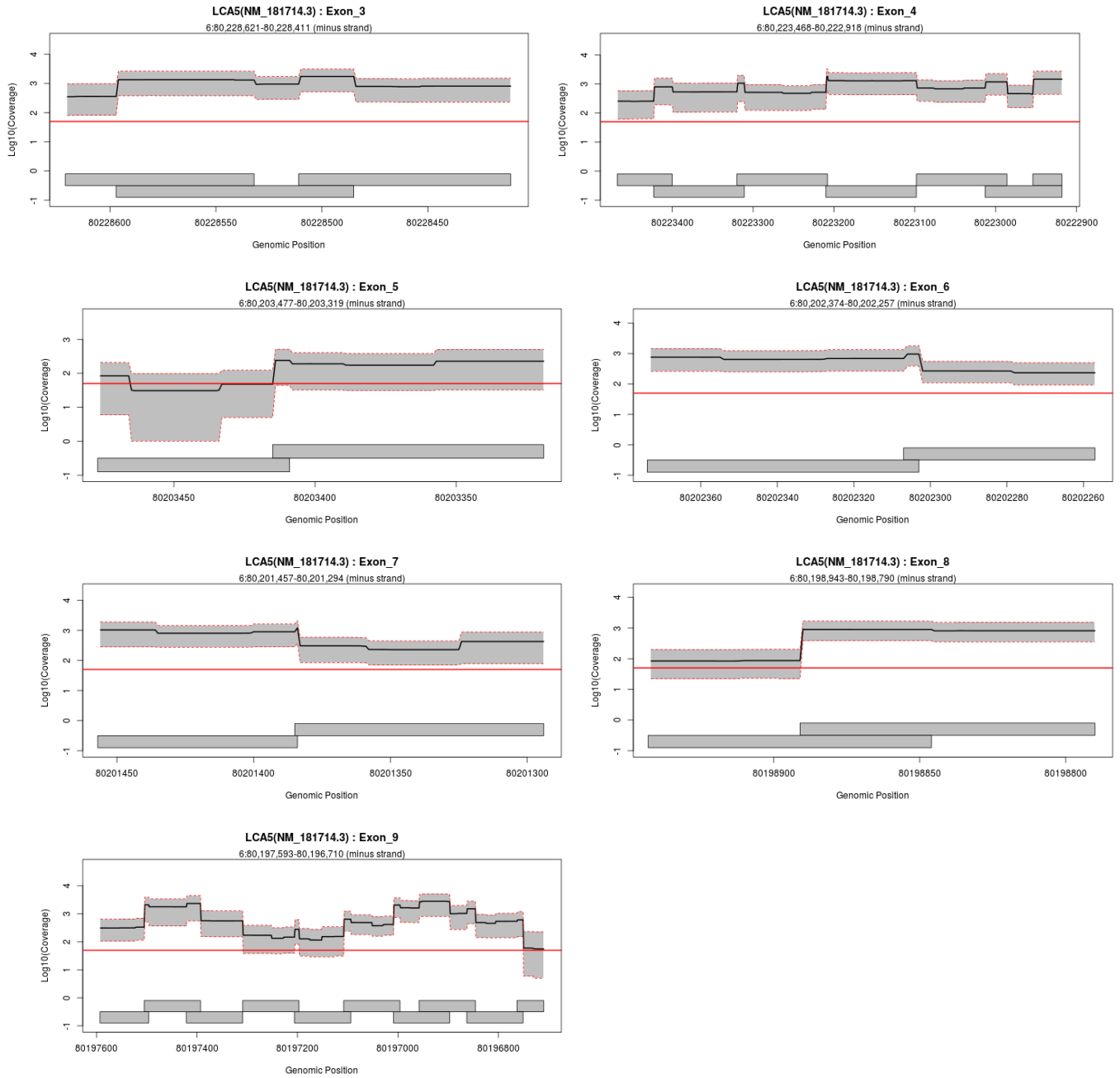




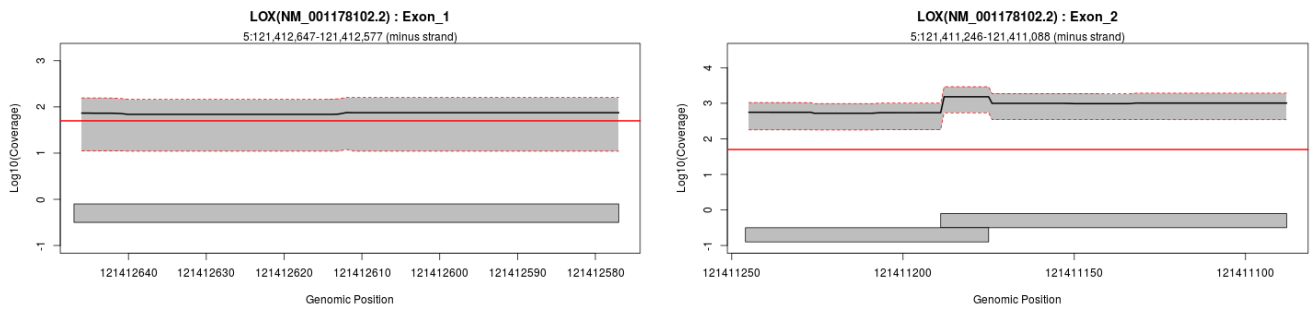
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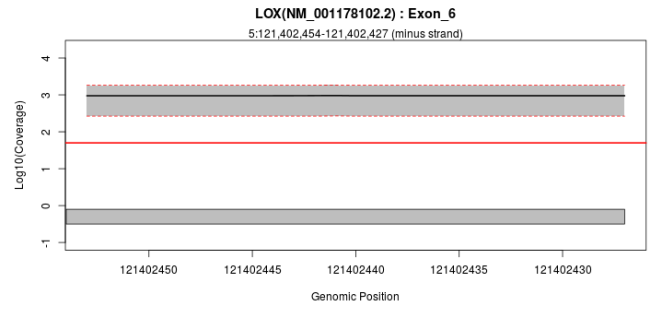
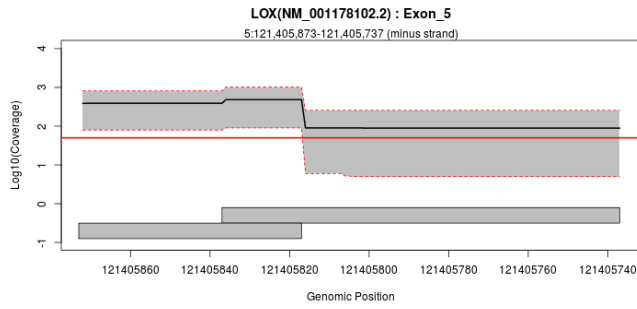
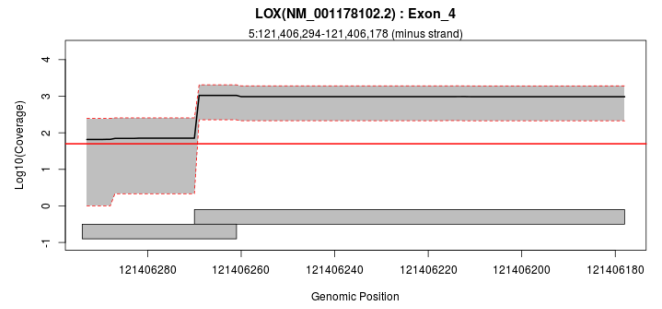
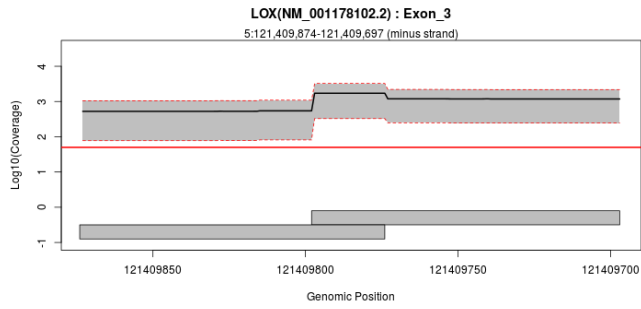


## Exon Overview: LCA5 : NM\_181714.3

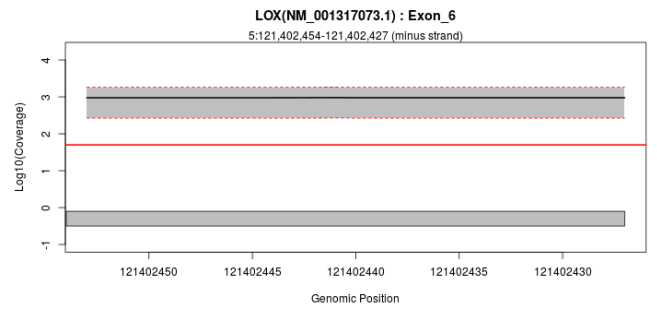
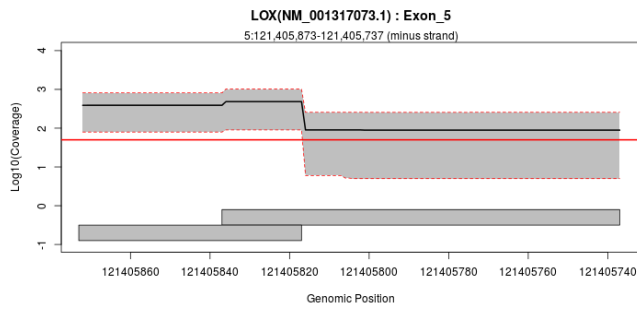
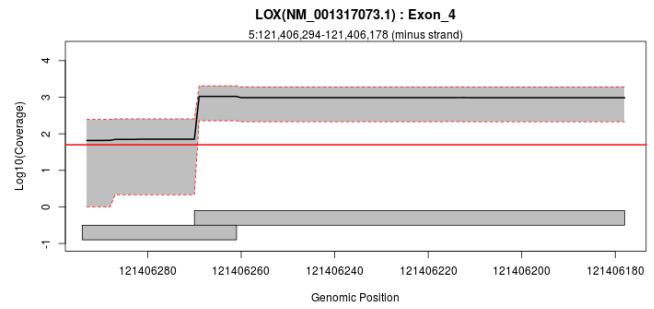
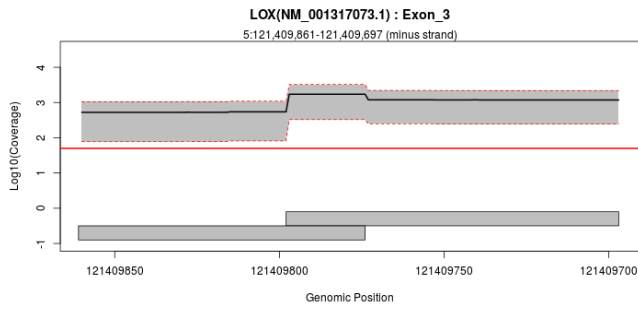


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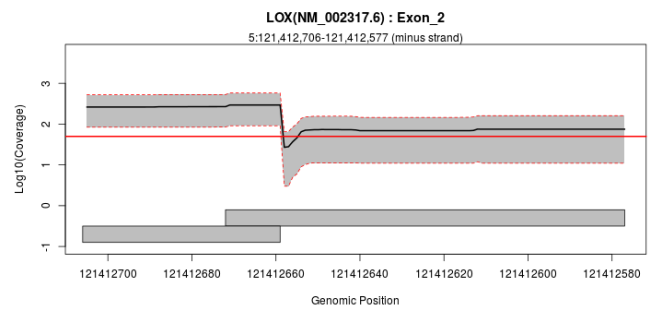
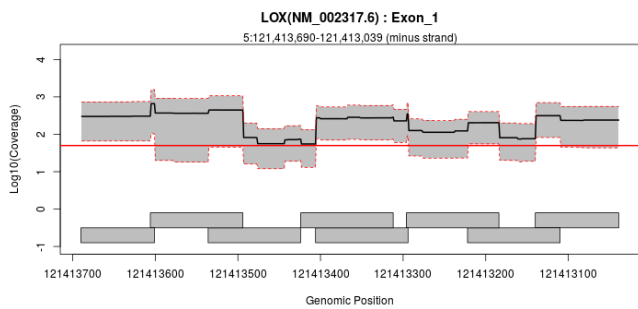




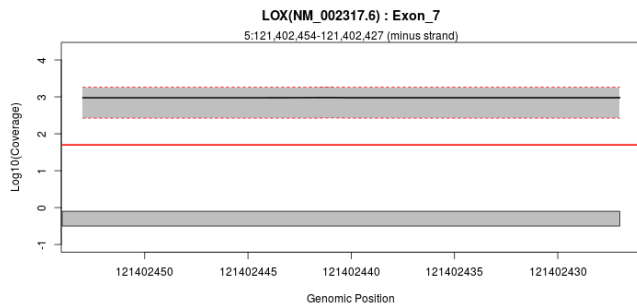
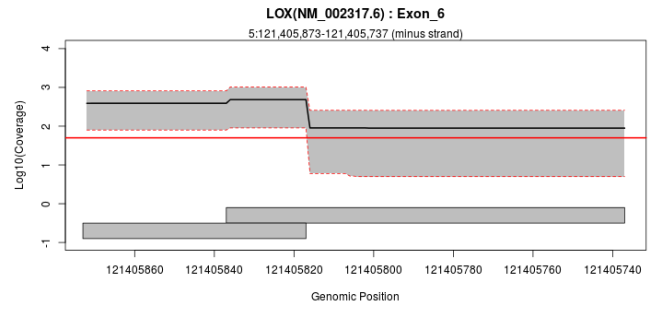
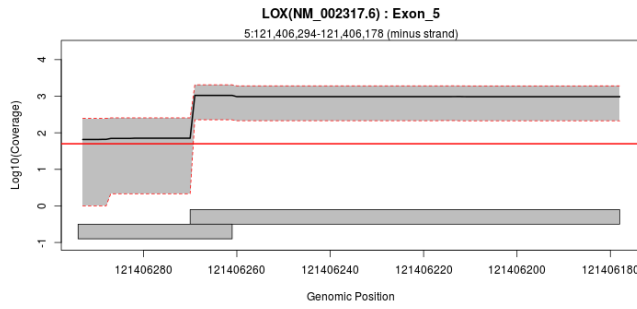
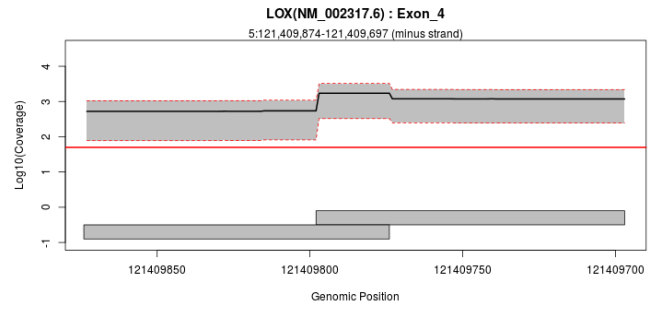
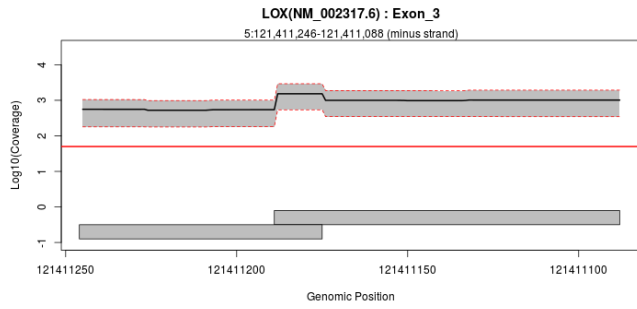
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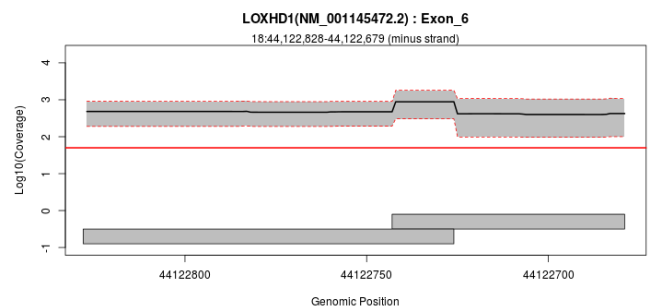
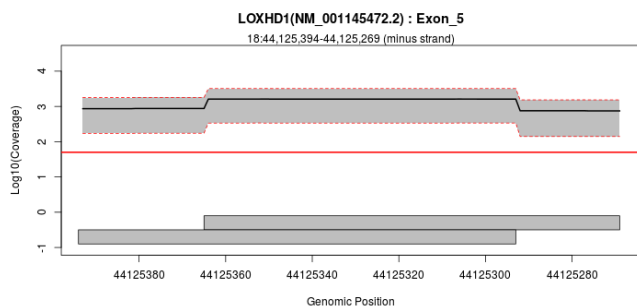
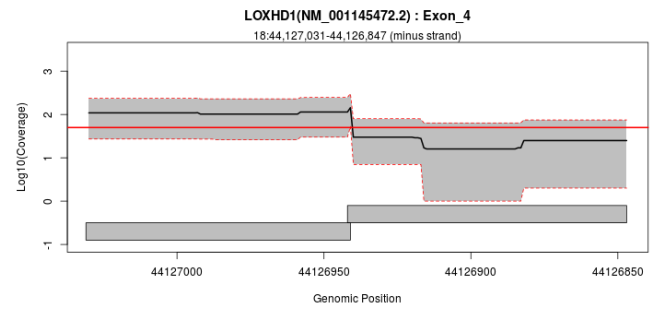
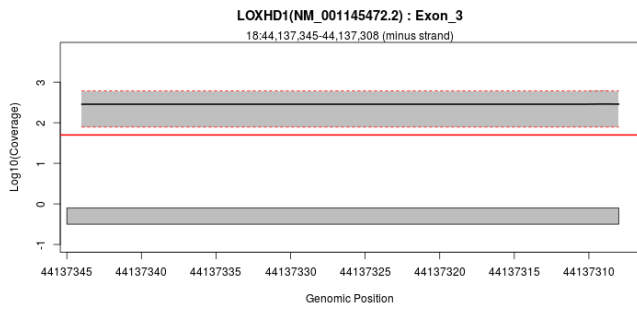
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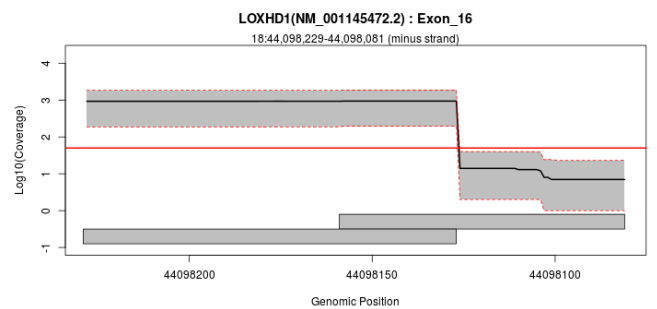
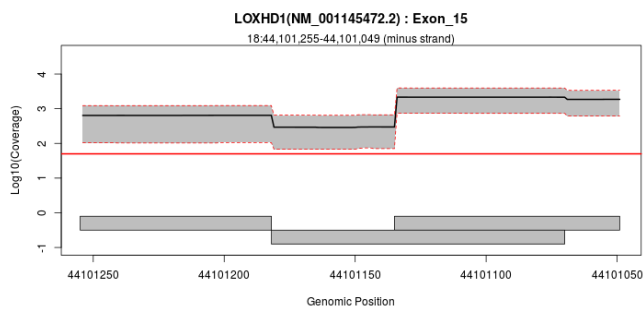
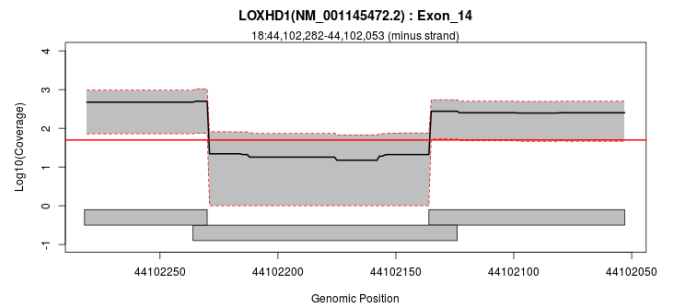
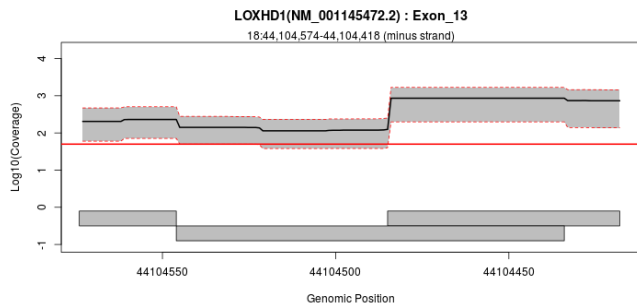
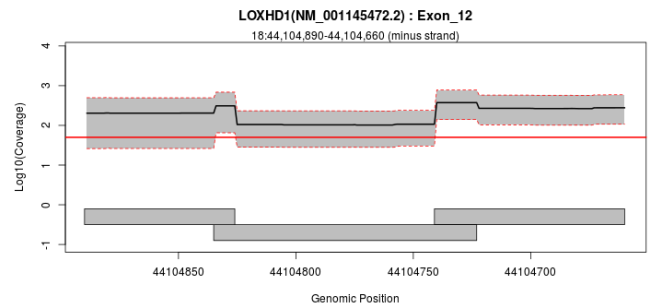
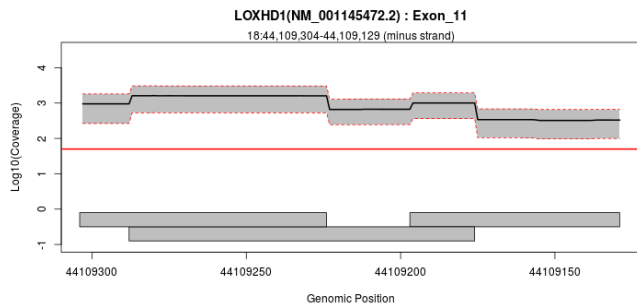
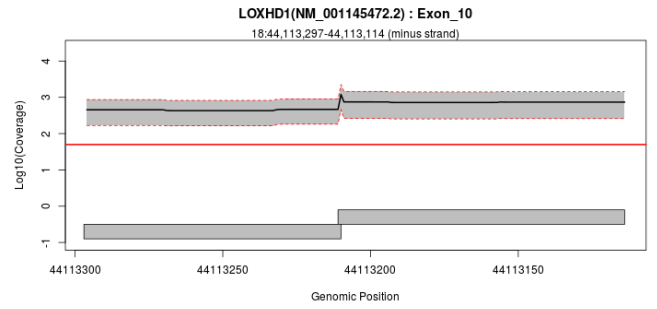
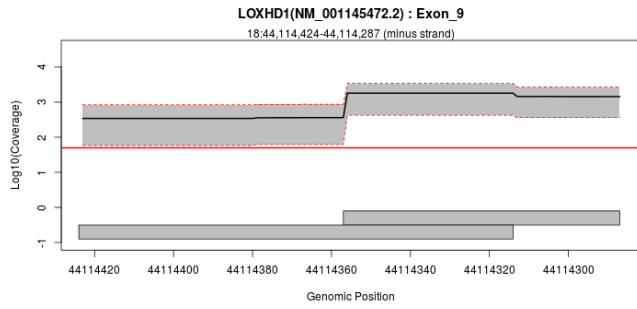
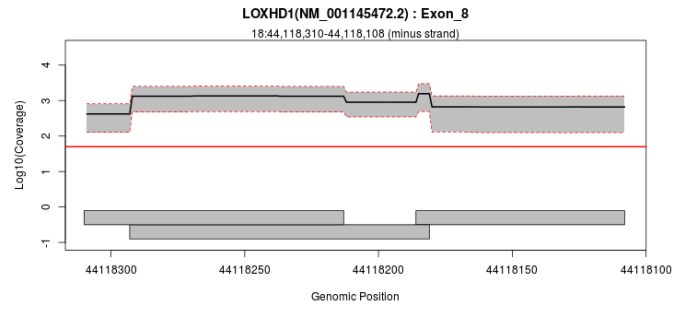
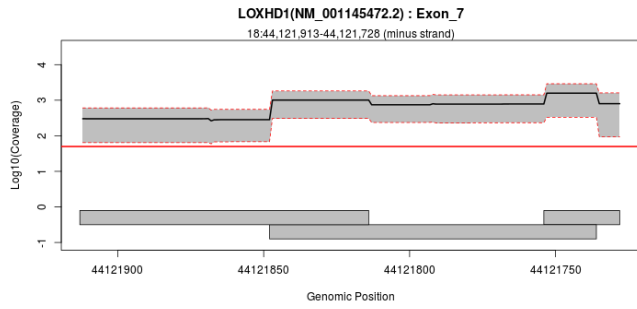


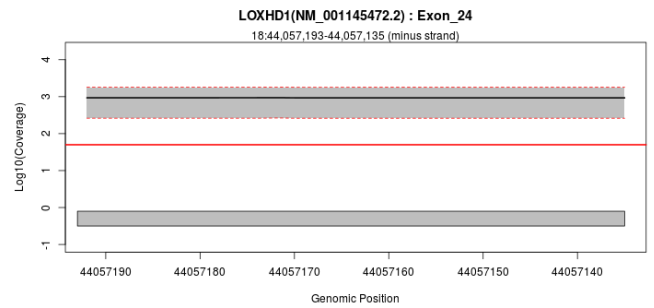
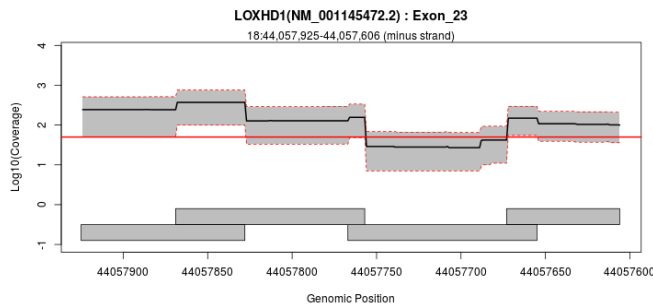
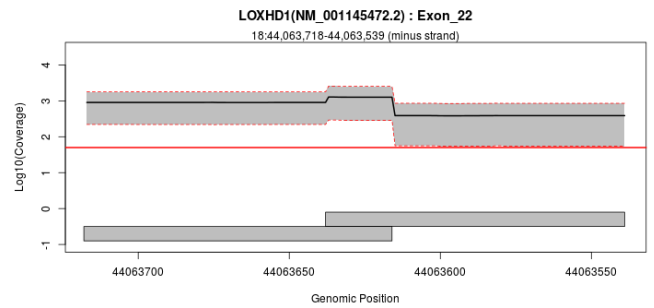
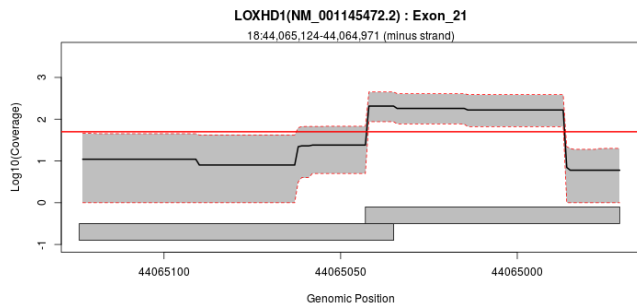
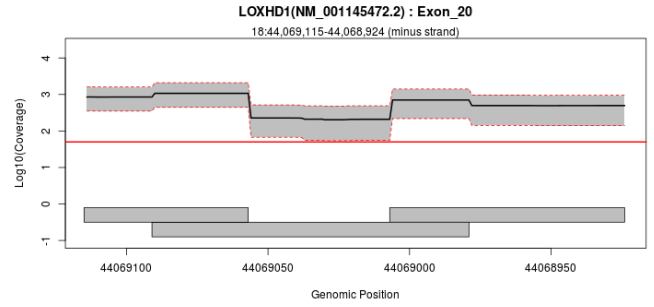
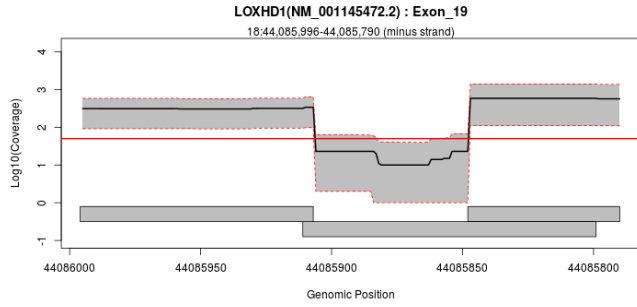
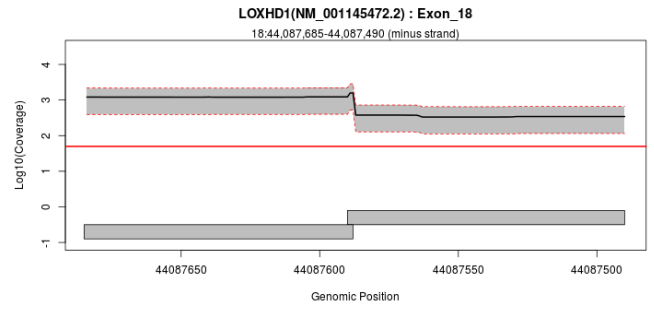
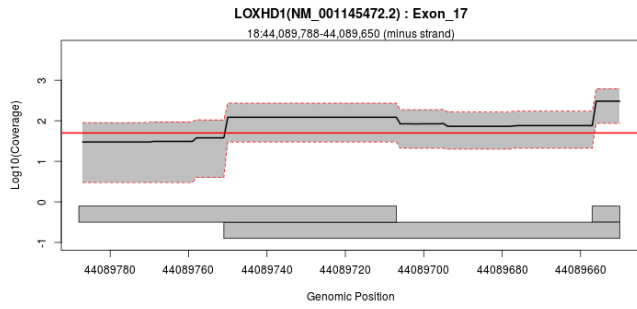




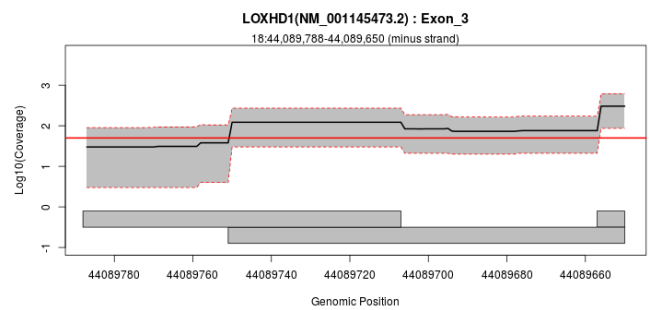
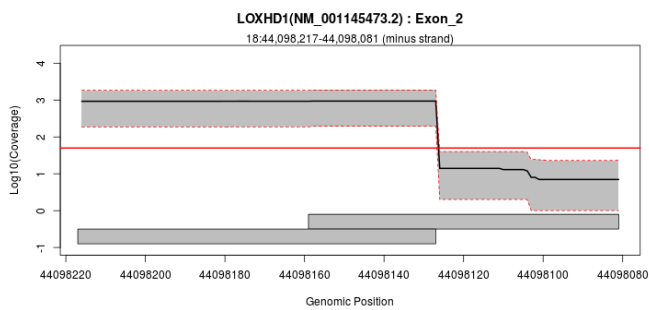
## Exon Overview: LOXHD1 : NM\_001145472.2

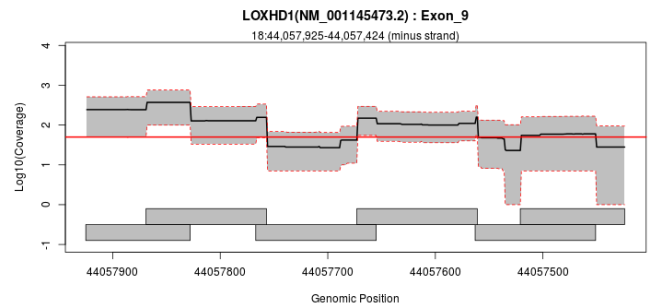
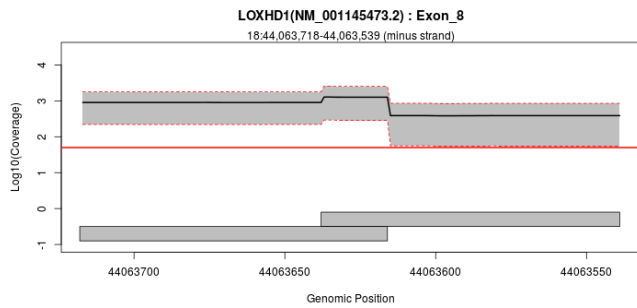
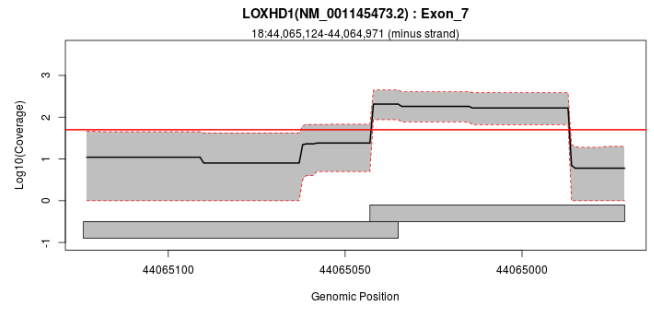
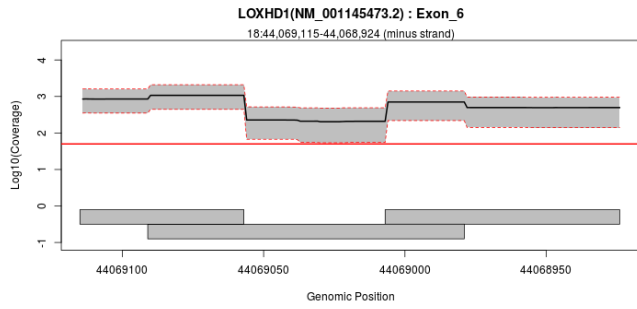
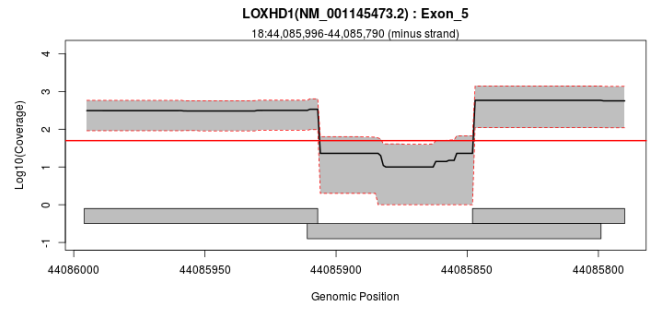
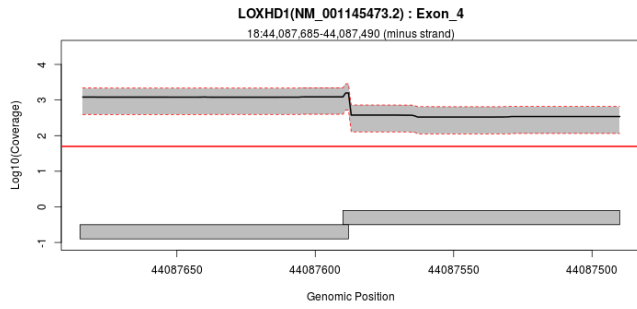




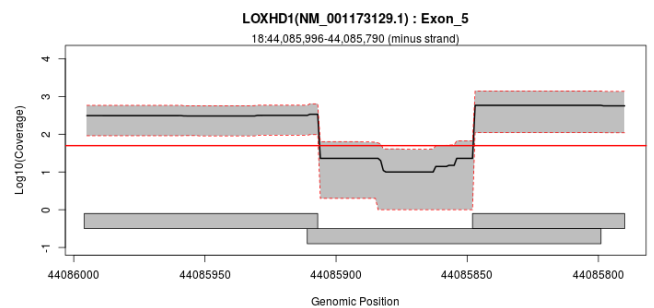
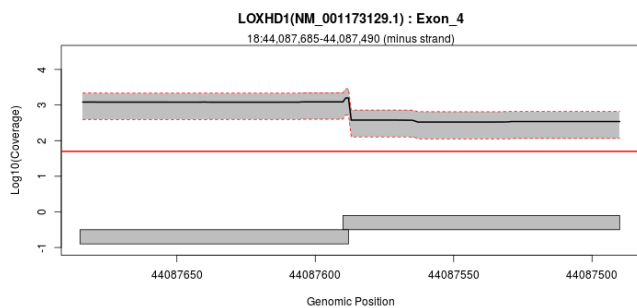
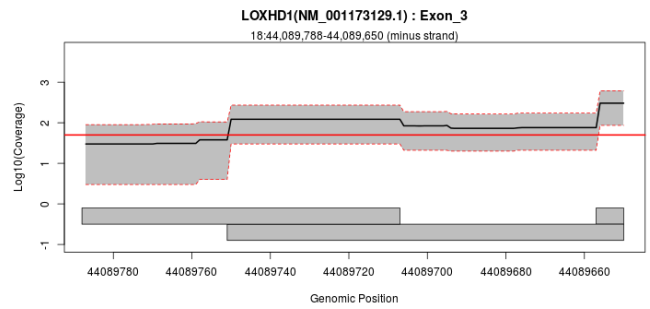
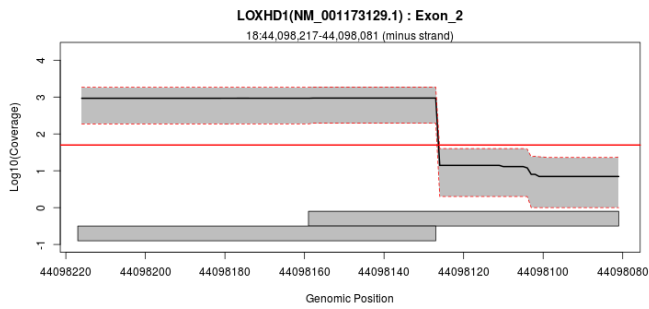


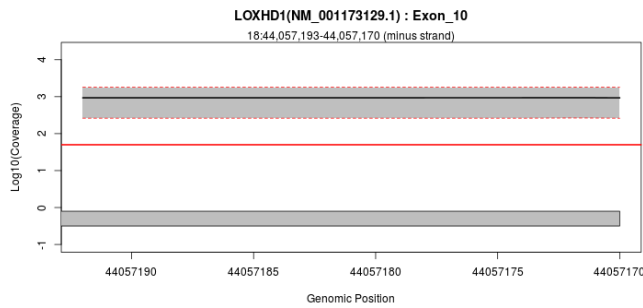
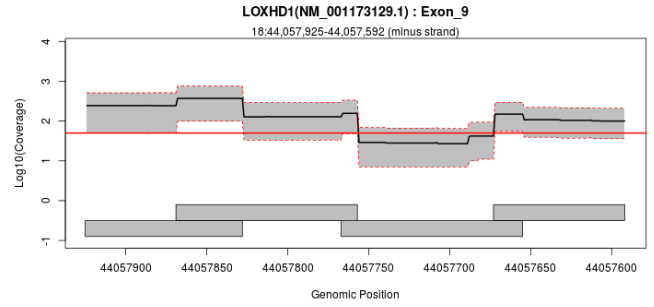
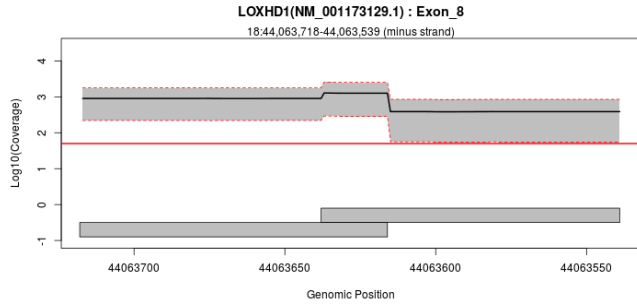
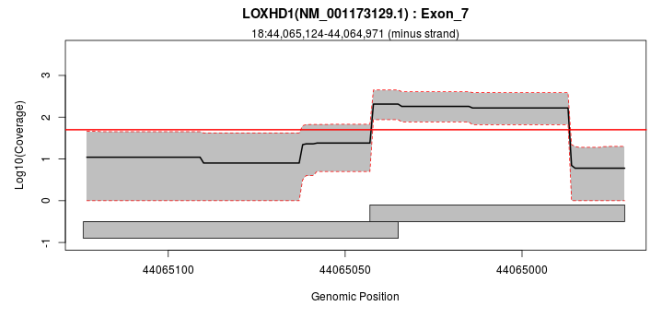
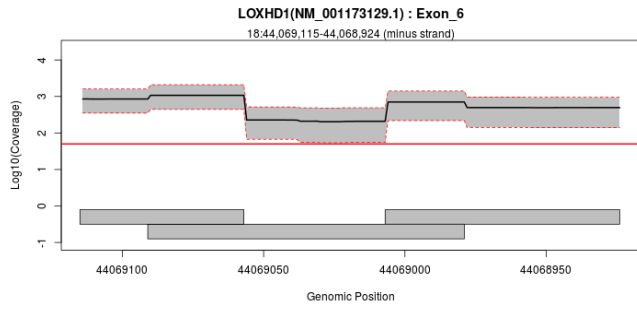
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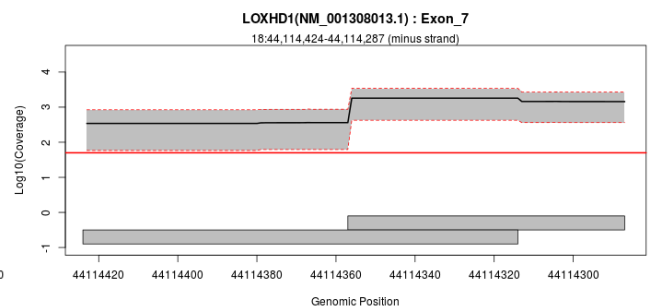
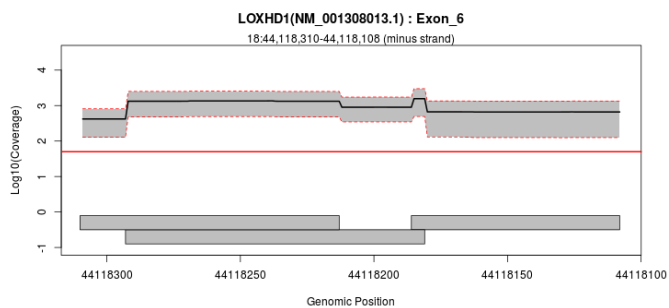
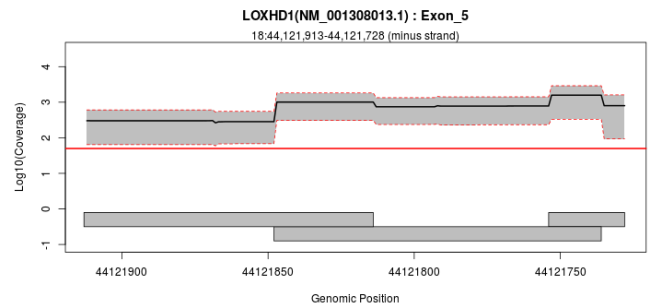
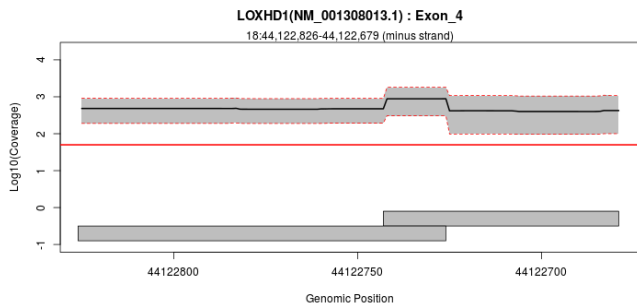


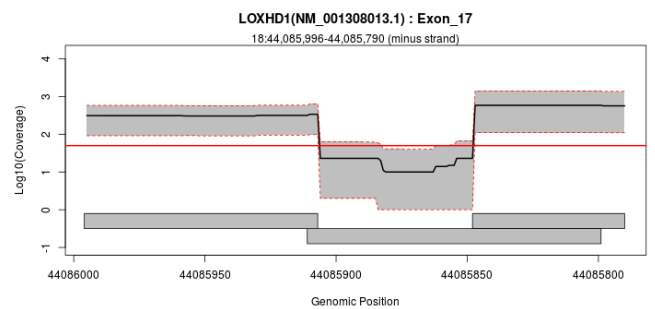
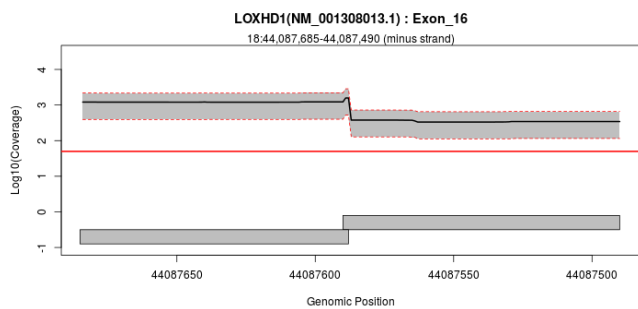
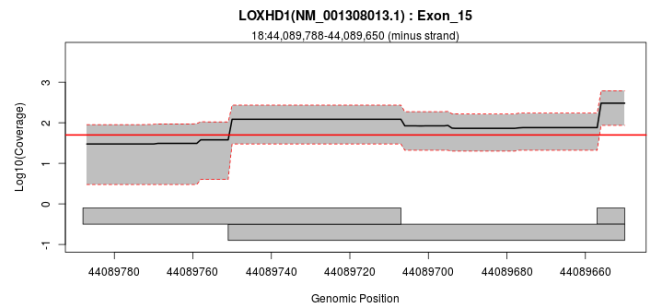
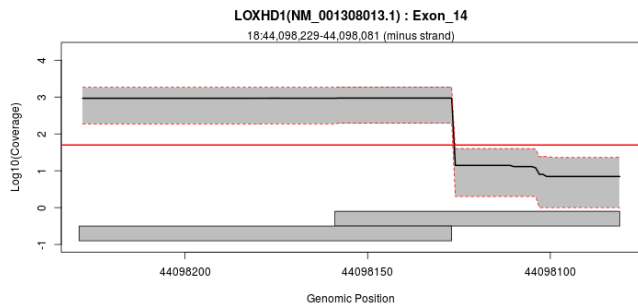
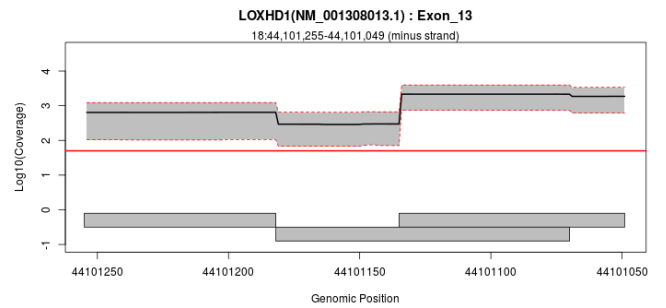
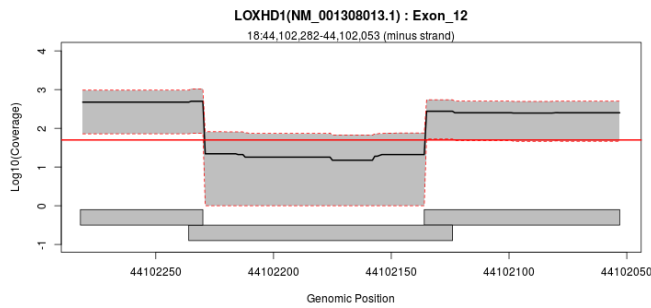
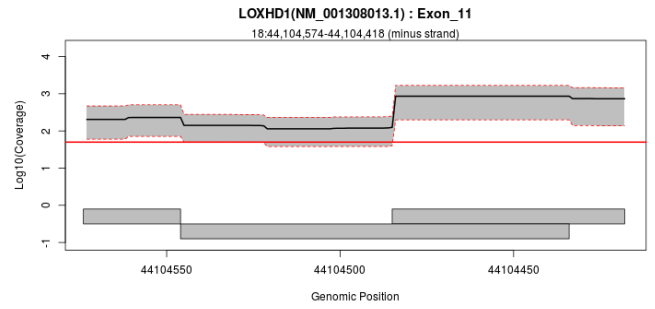
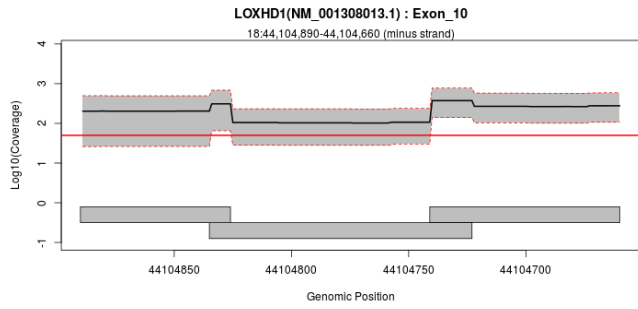
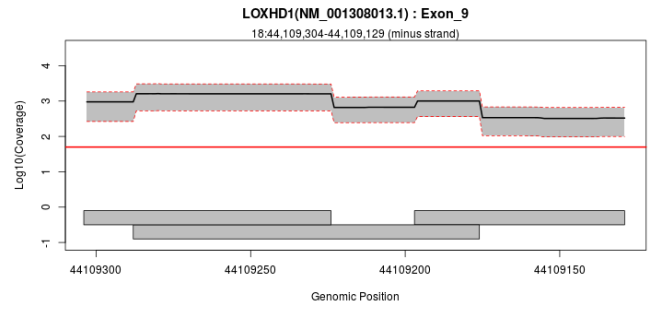
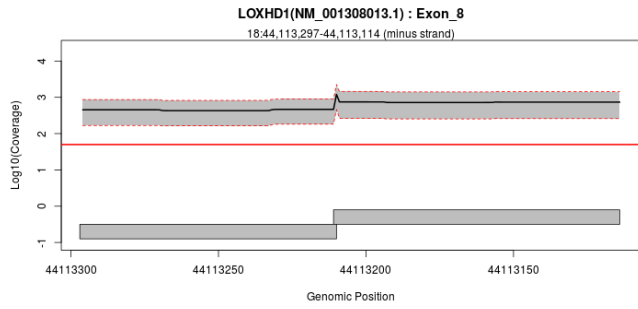
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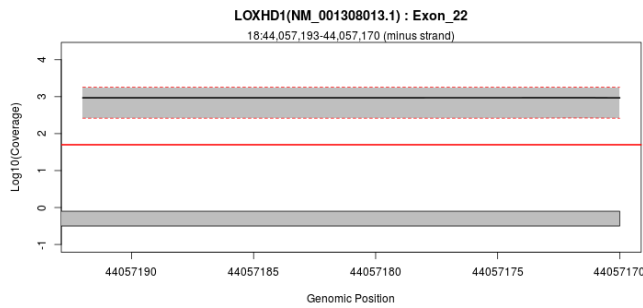
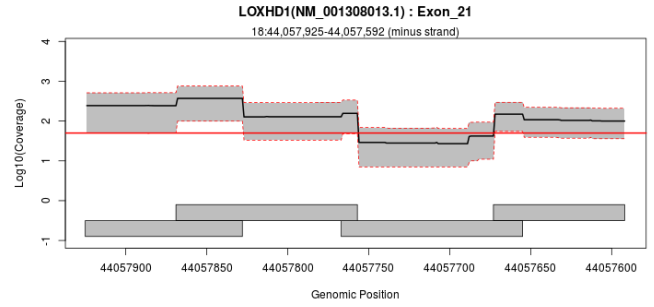
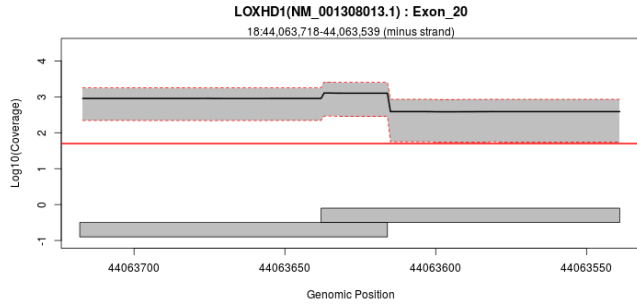
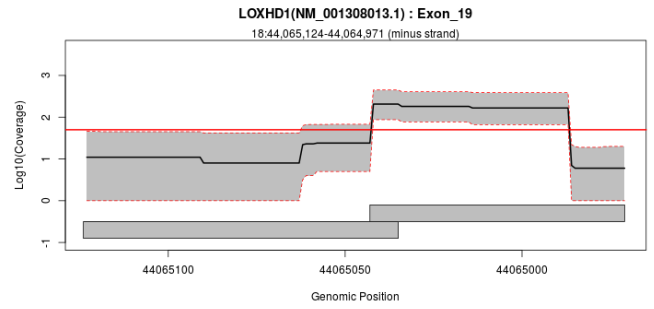
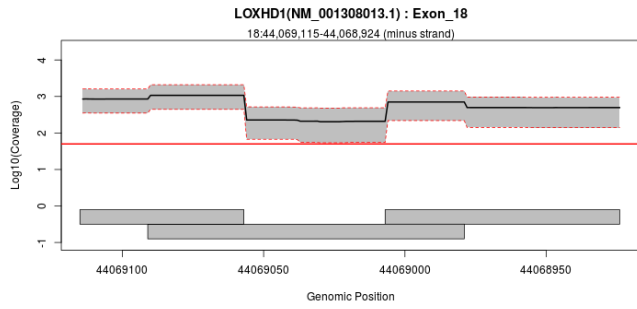




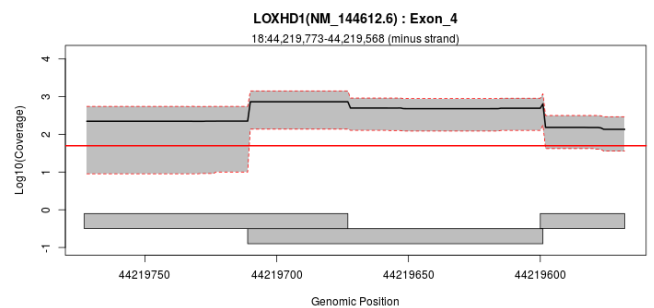
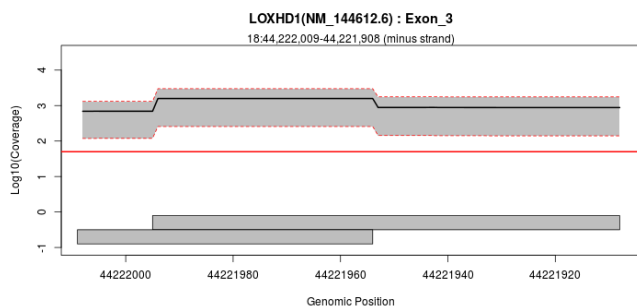
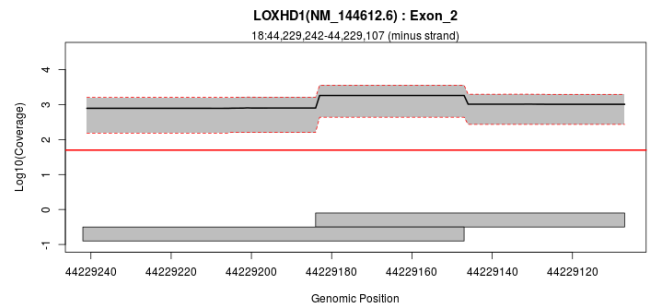
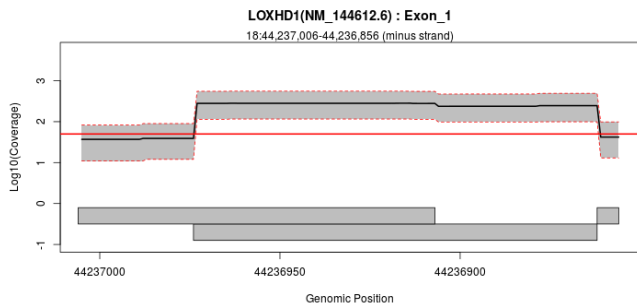
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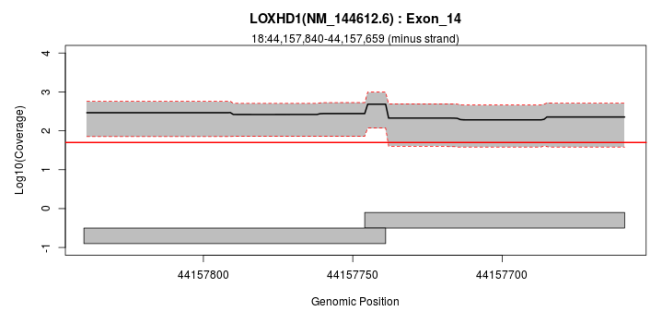
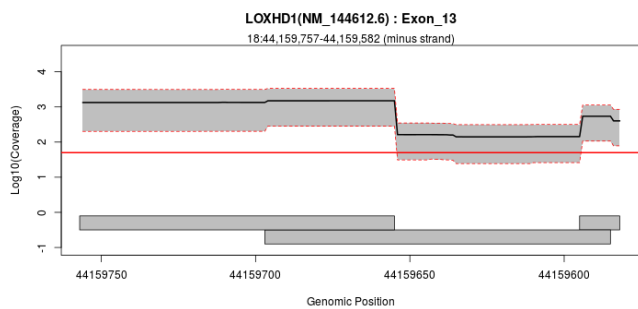
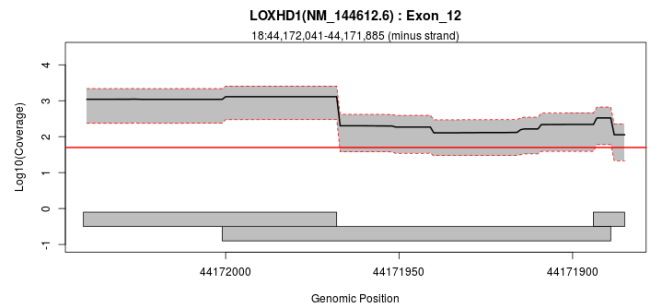
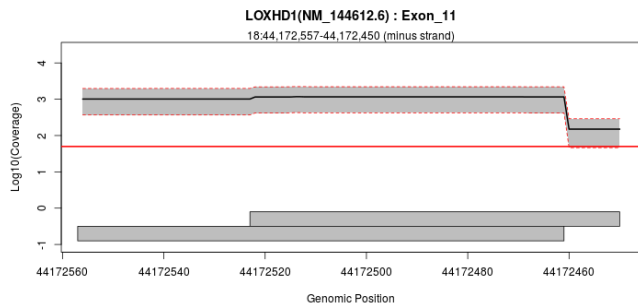
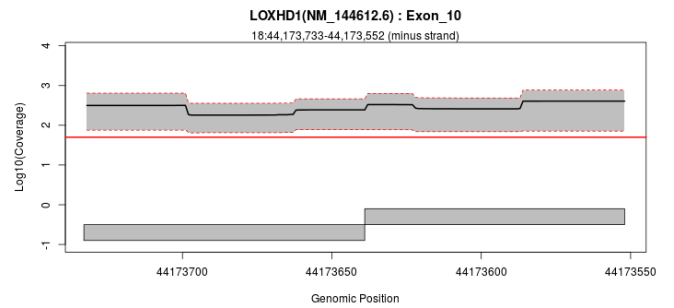
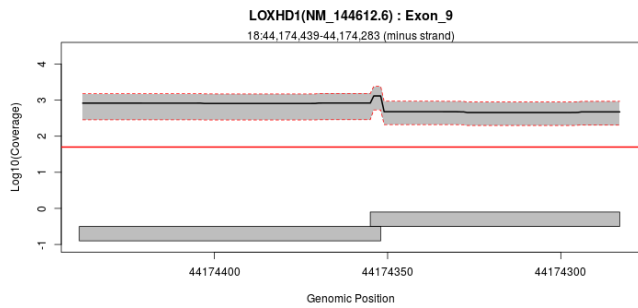
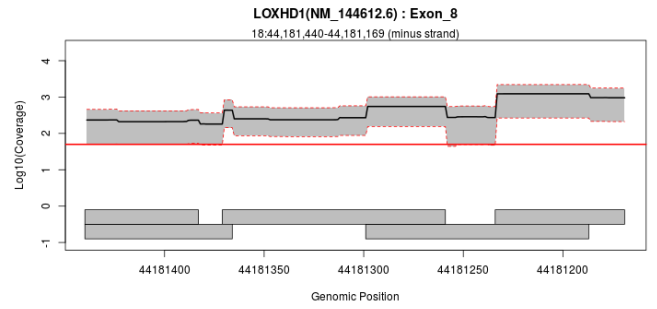
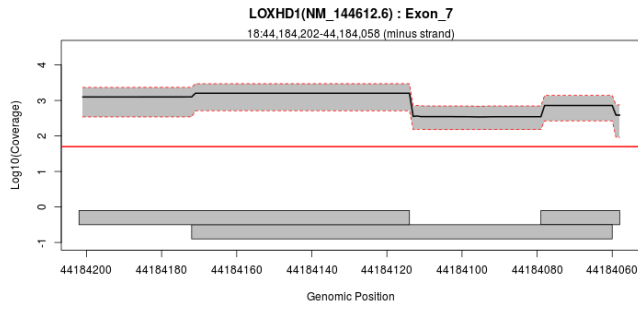
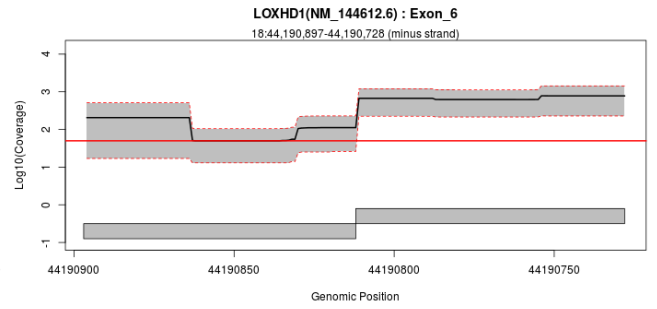
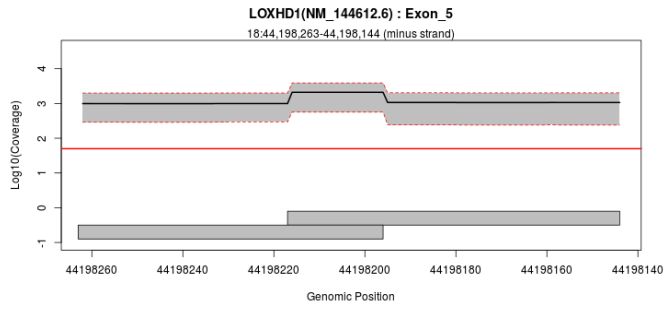




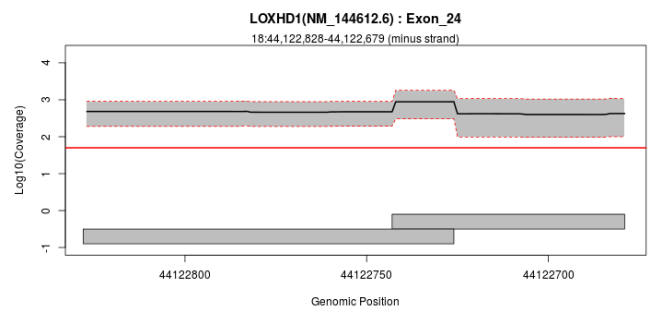
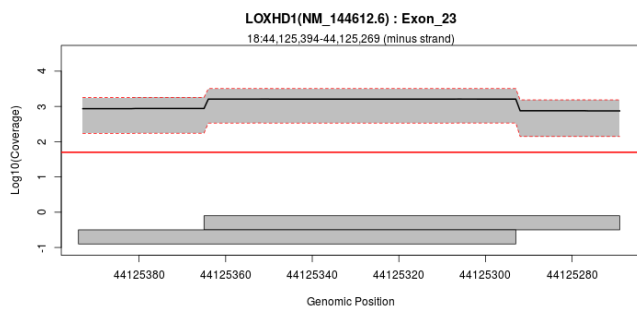
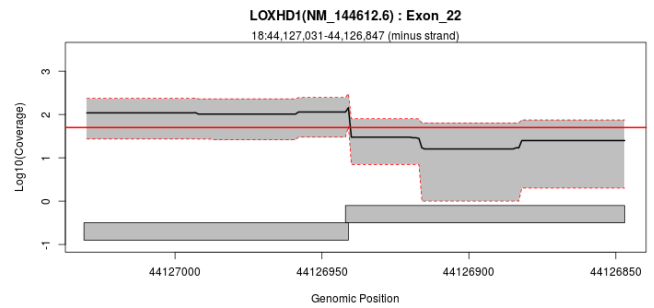
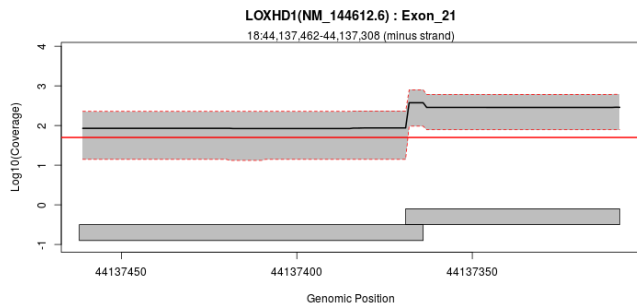
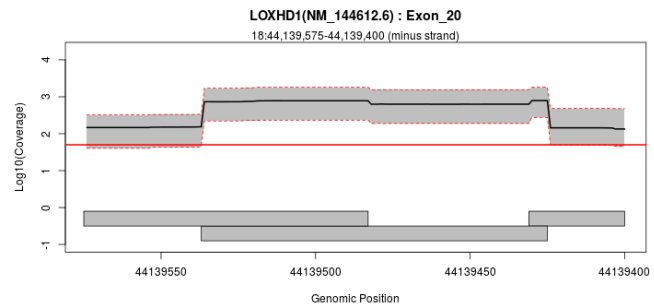
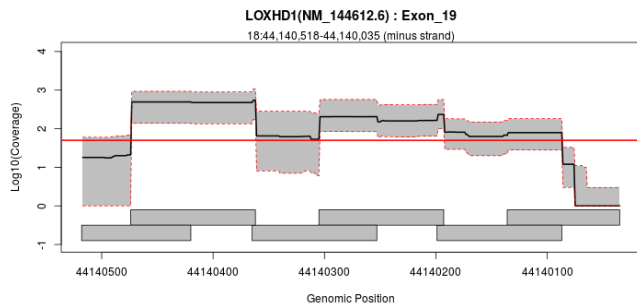
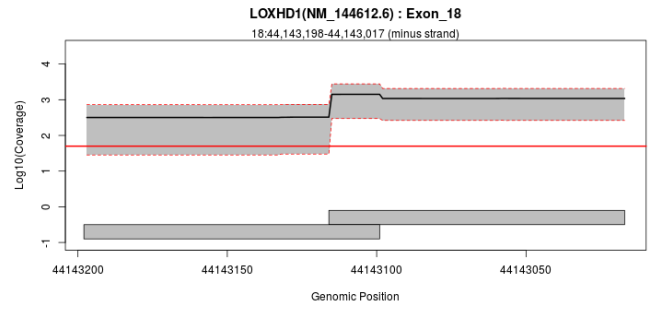
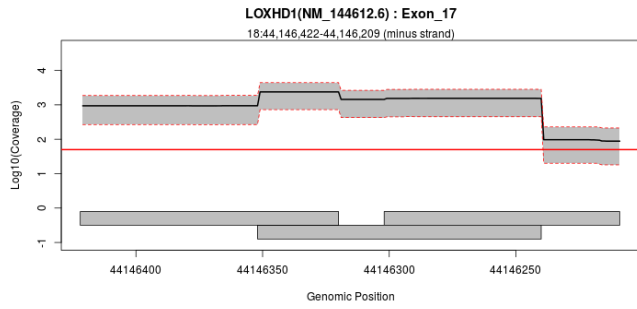
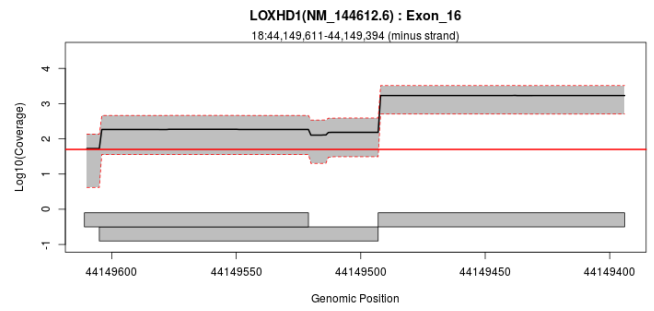
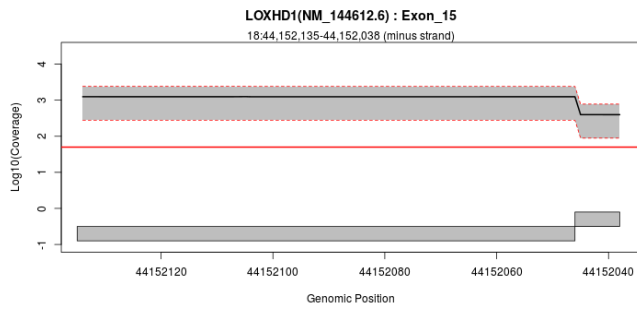


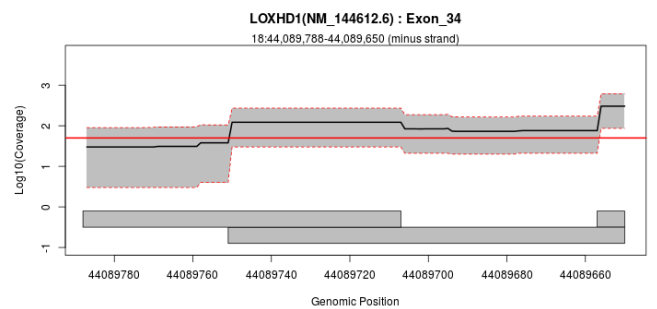
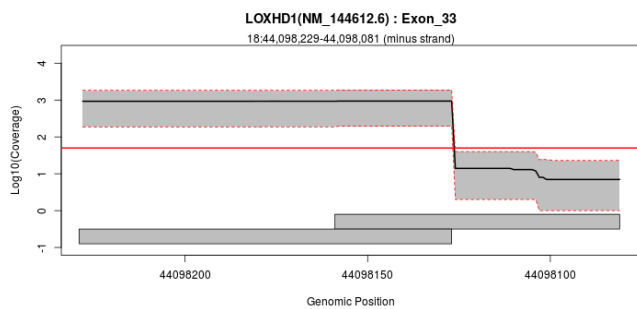
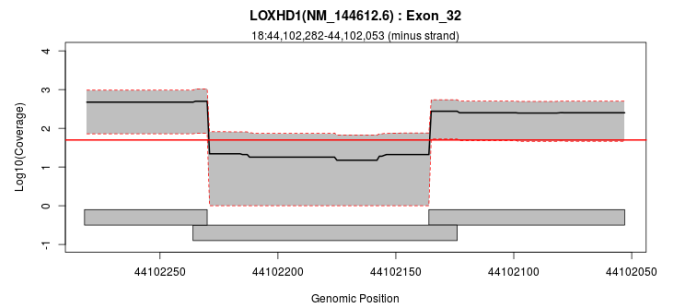
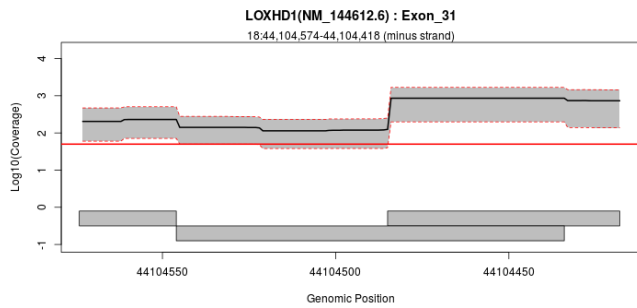
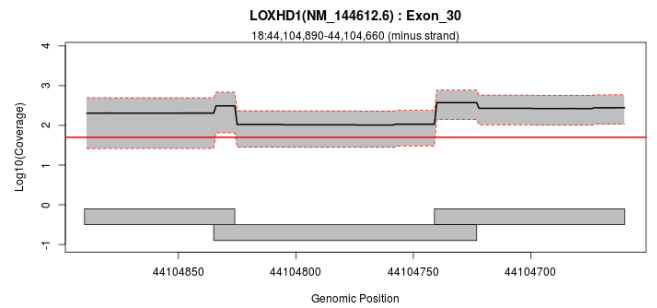
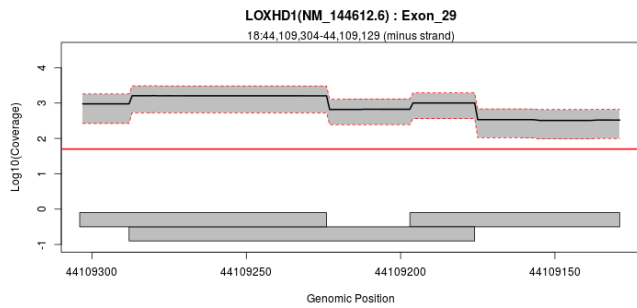
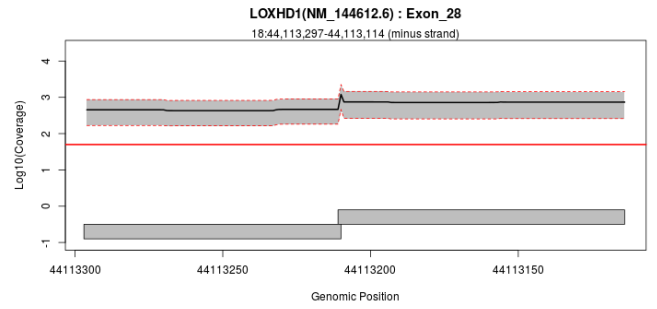
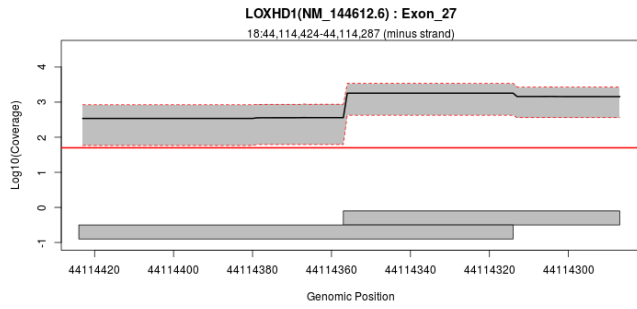
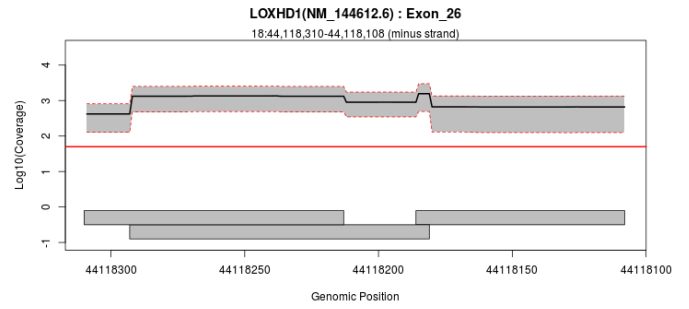
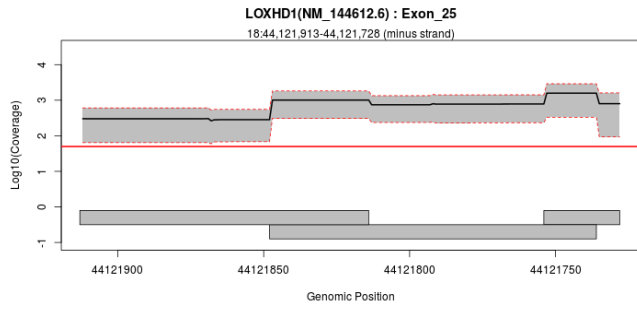
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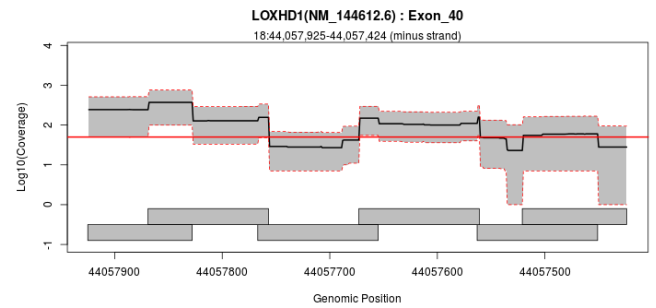
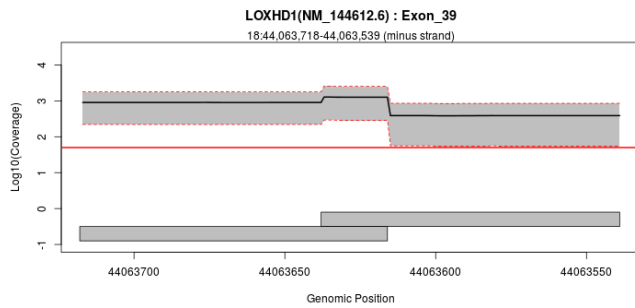
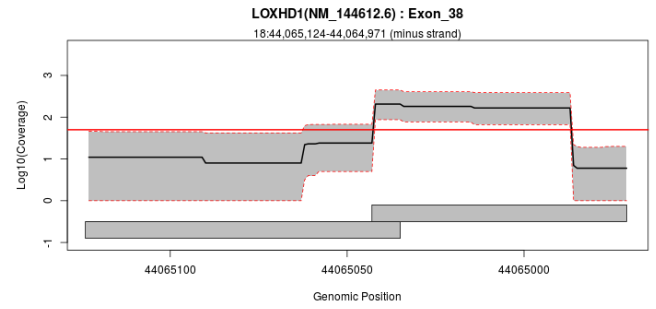
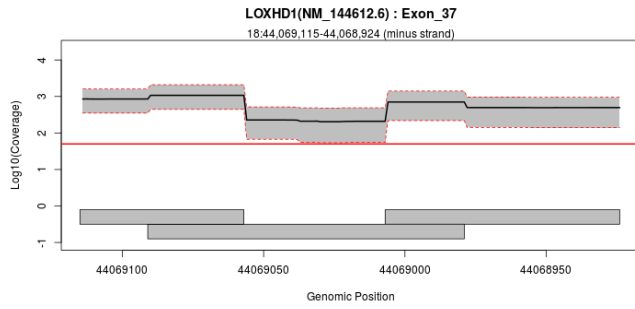
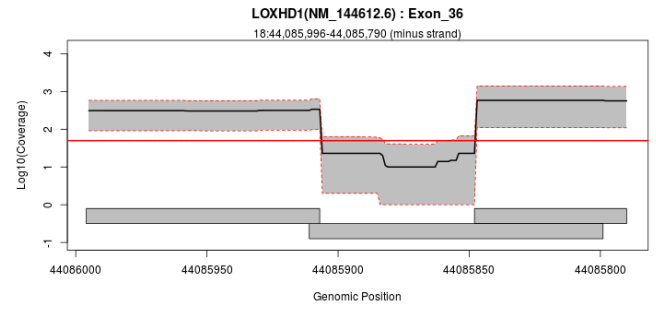
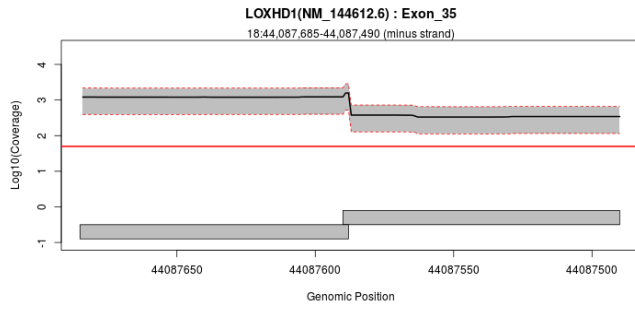




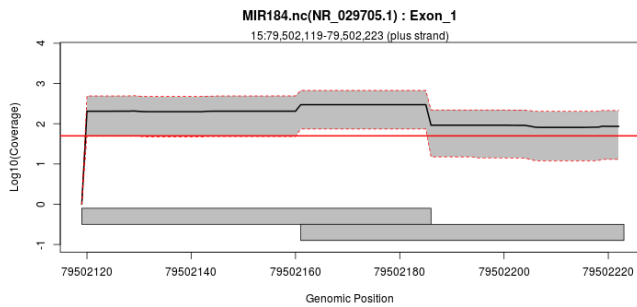




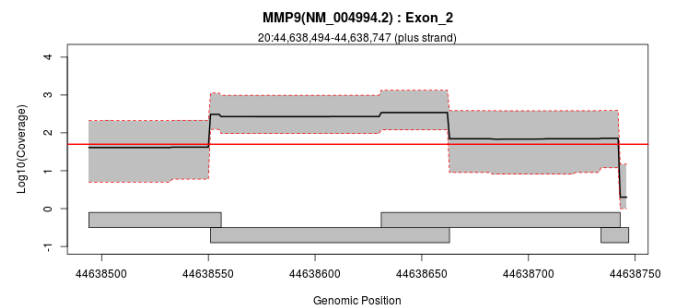
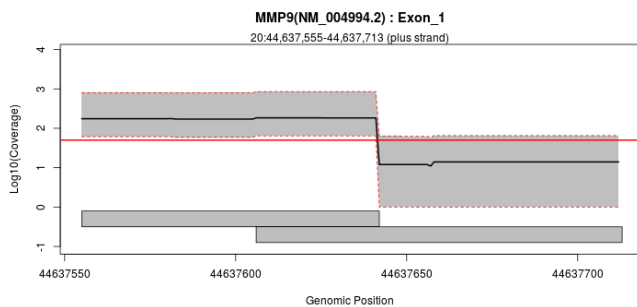


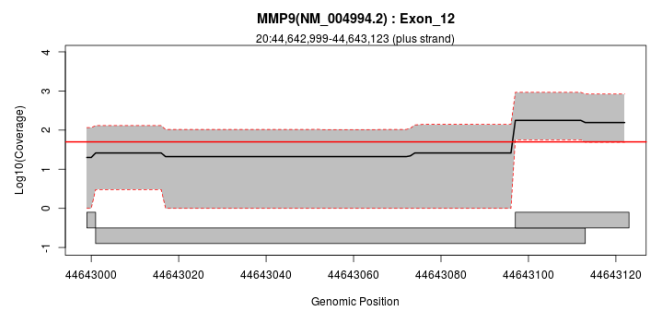
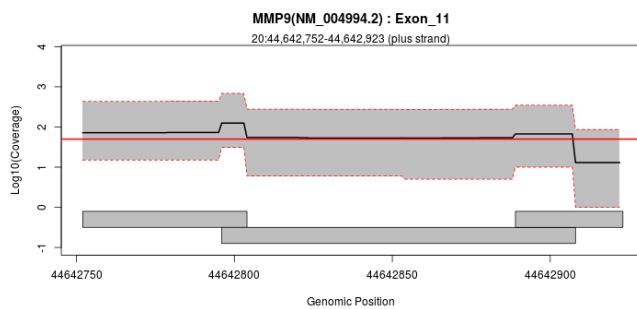
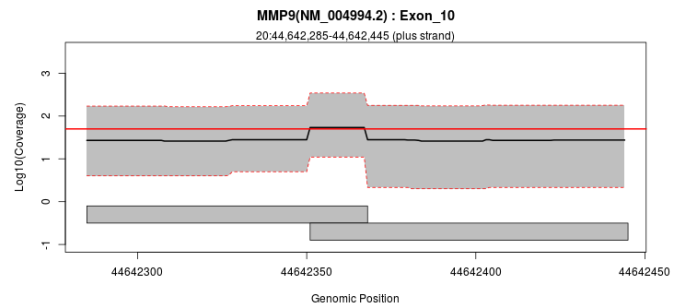
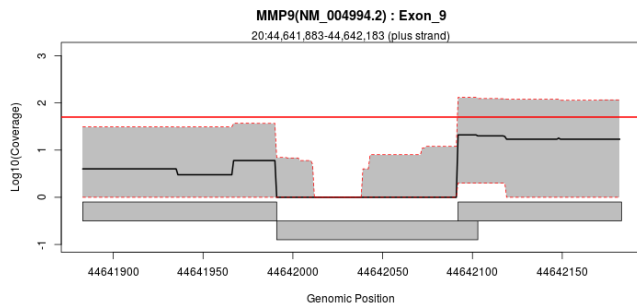
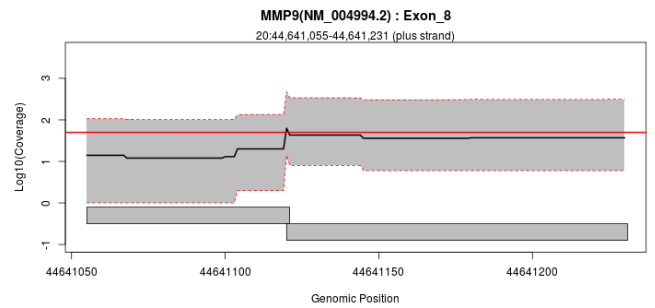
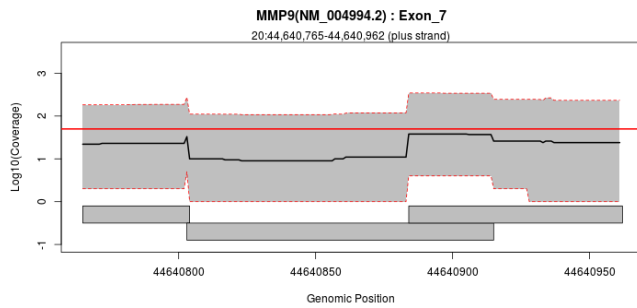
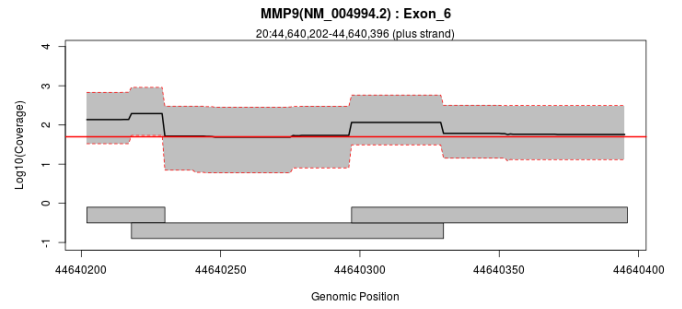
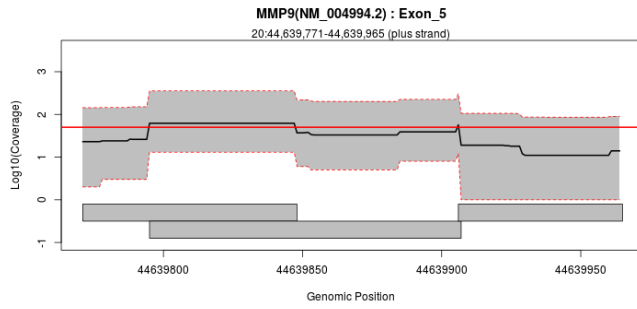
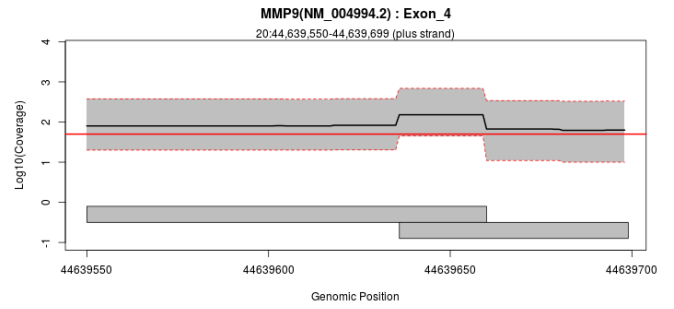
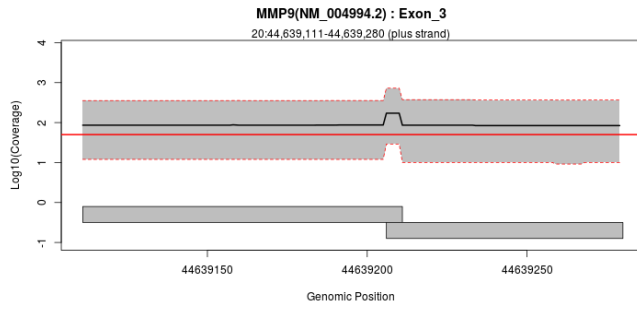


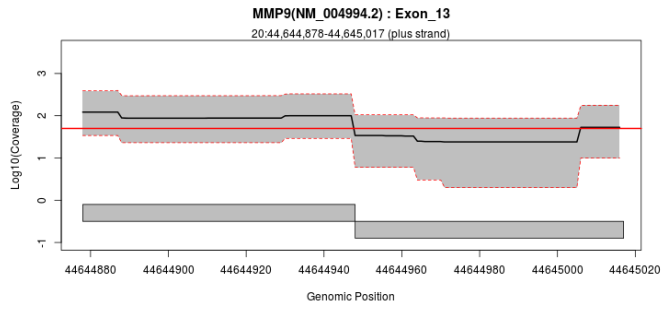
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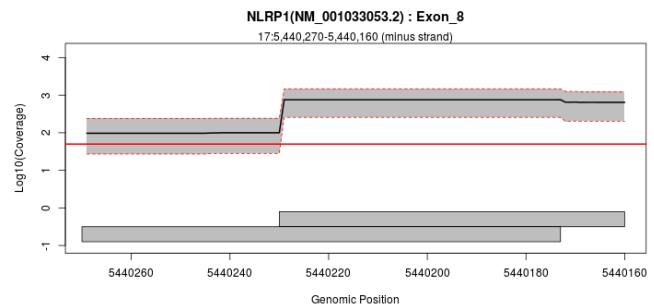
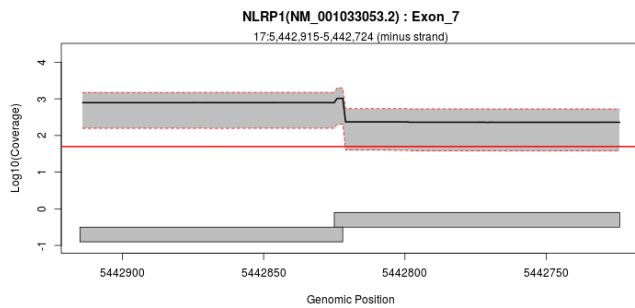
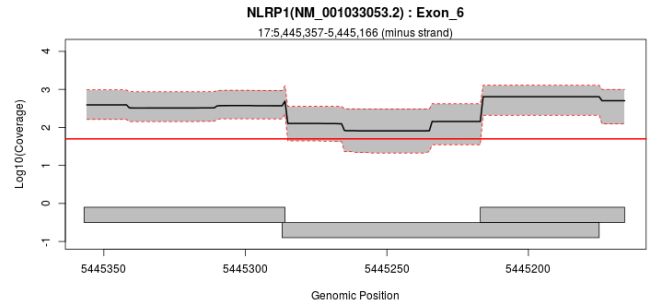
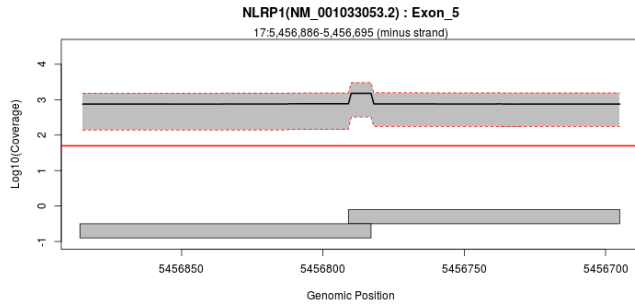
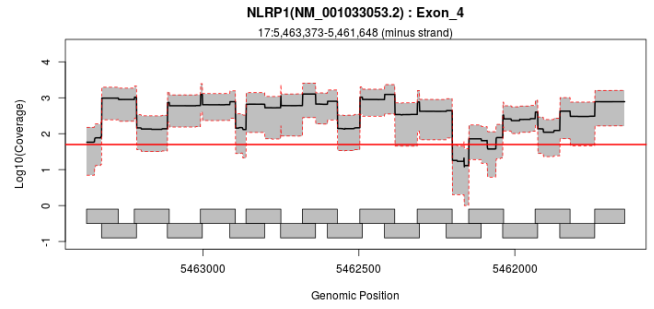
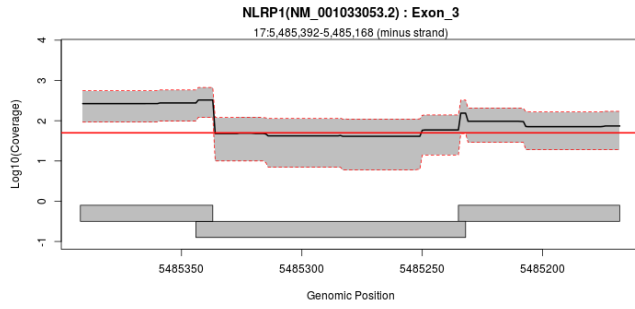
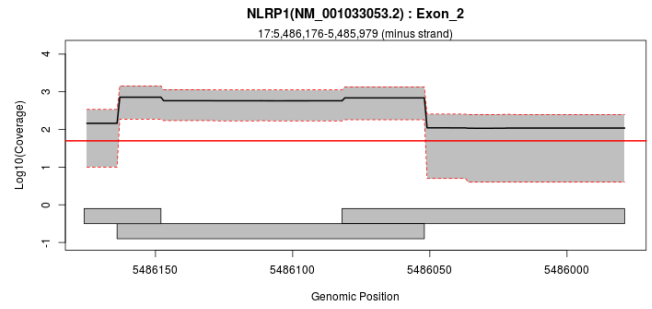
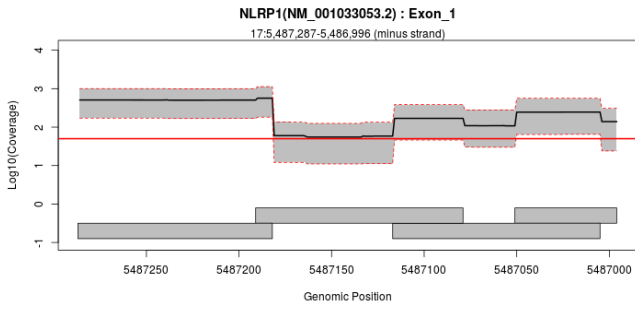
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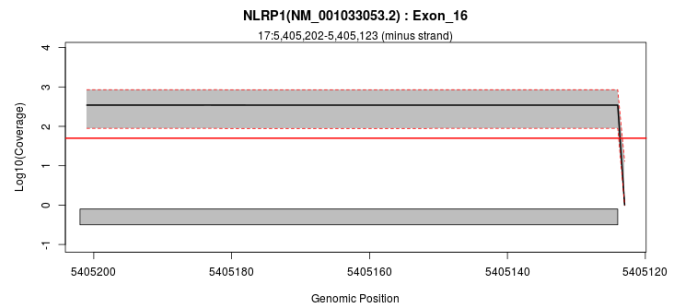
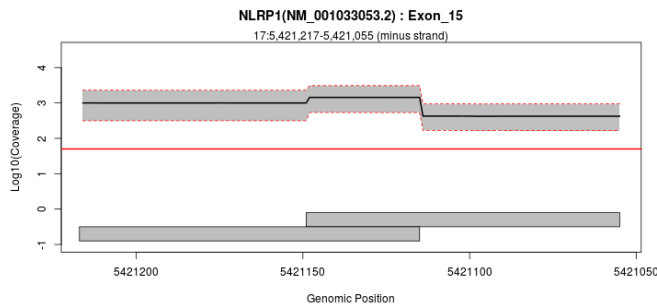
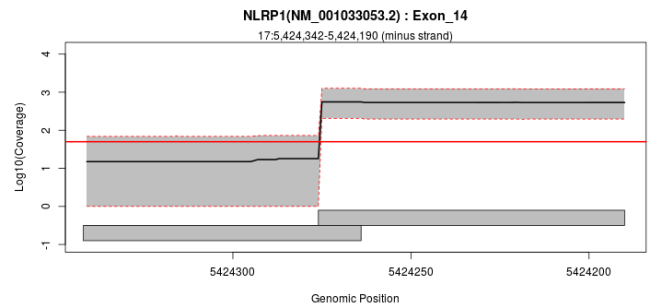
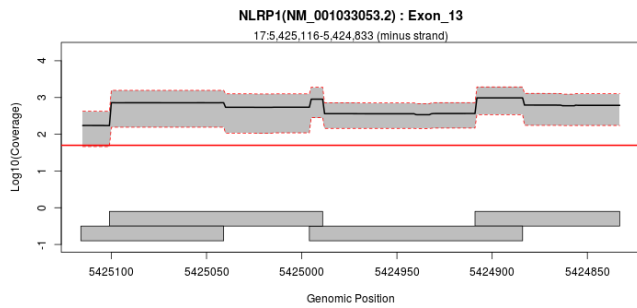
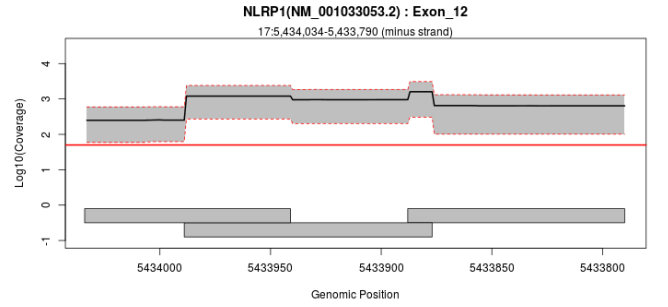
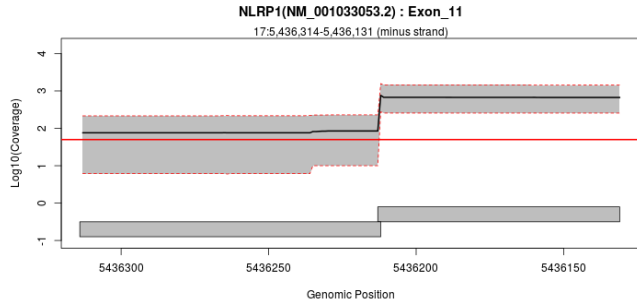
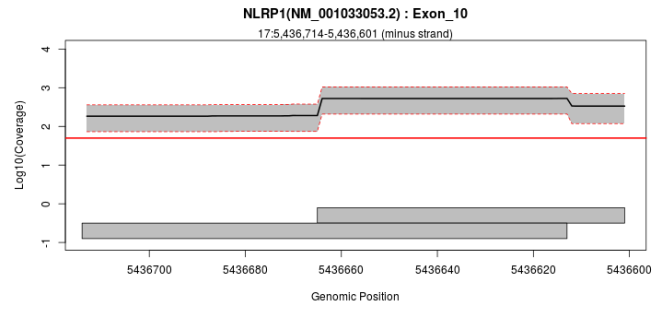
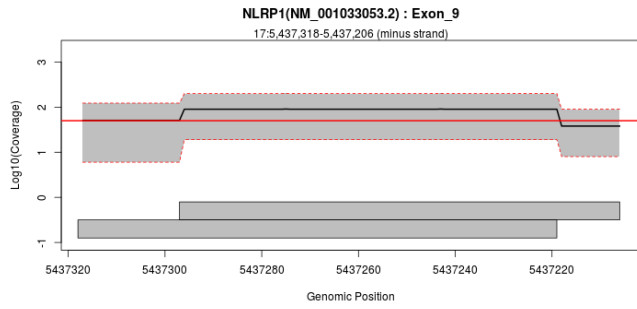




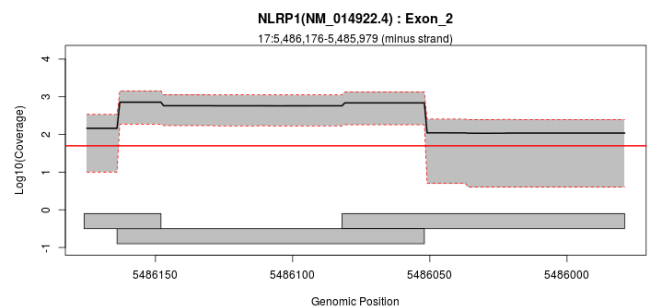
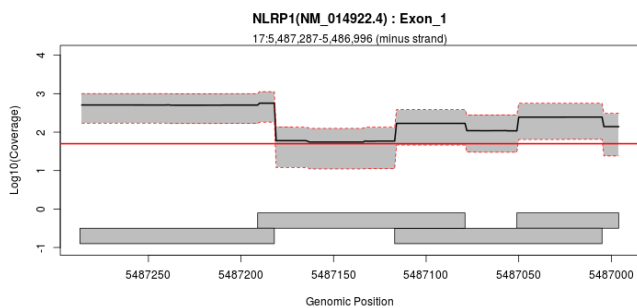


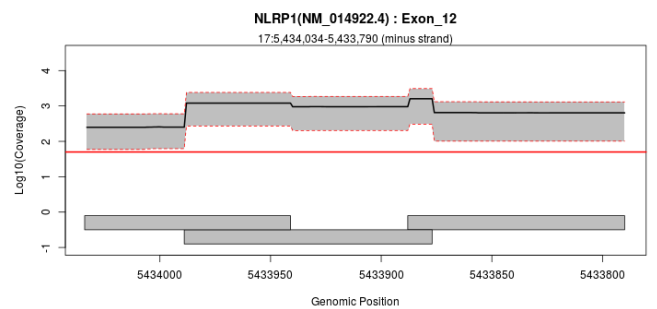
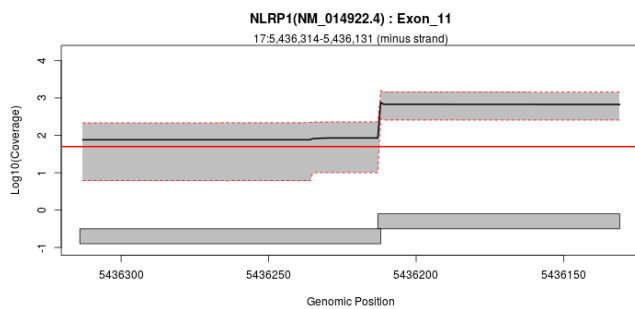
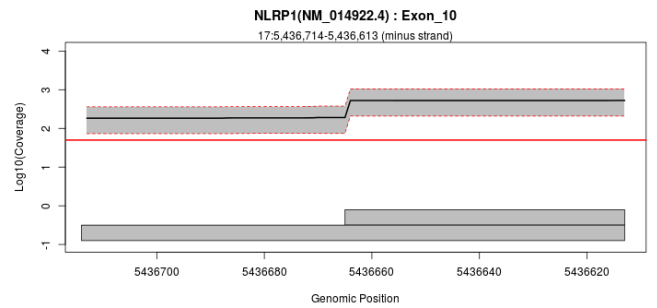
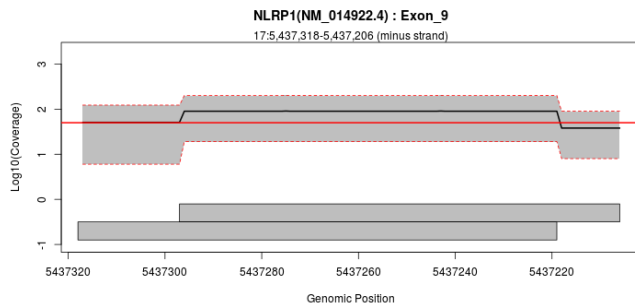
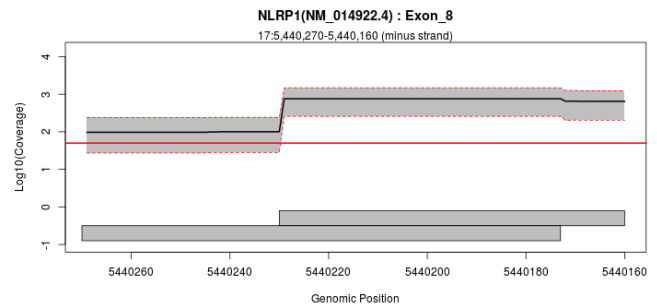
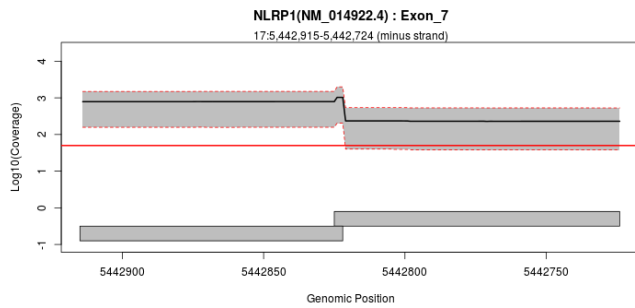
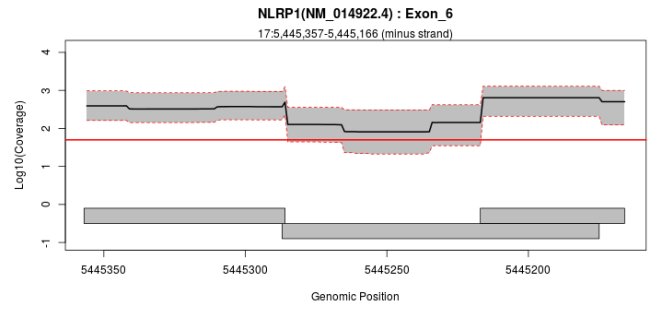
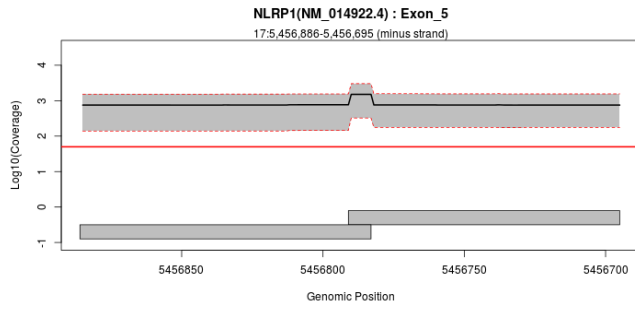
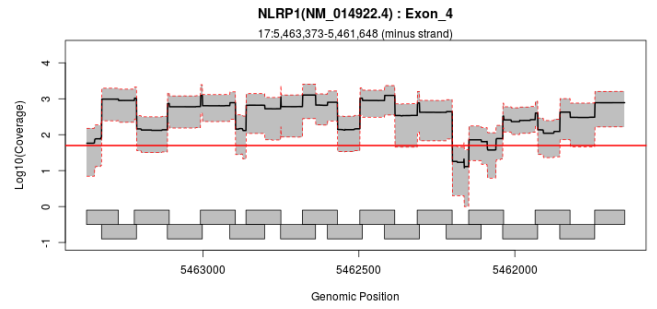
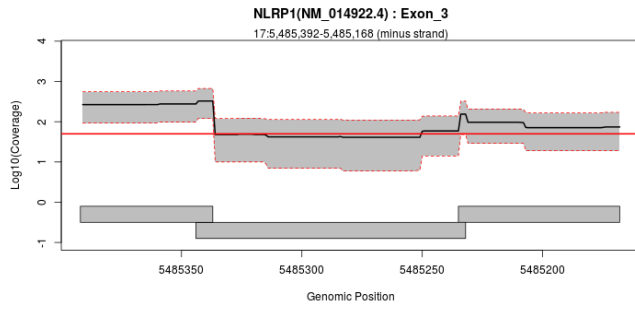
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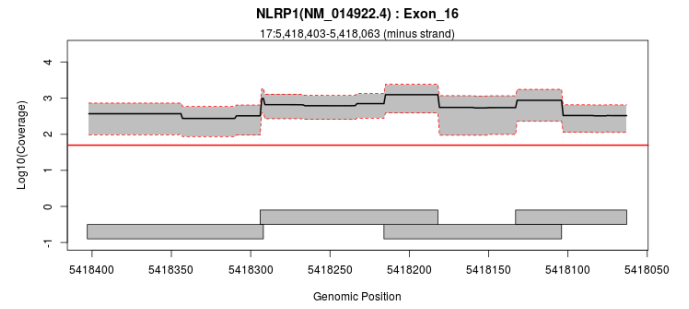
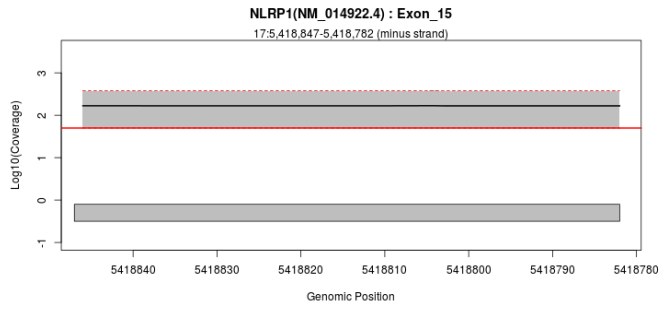
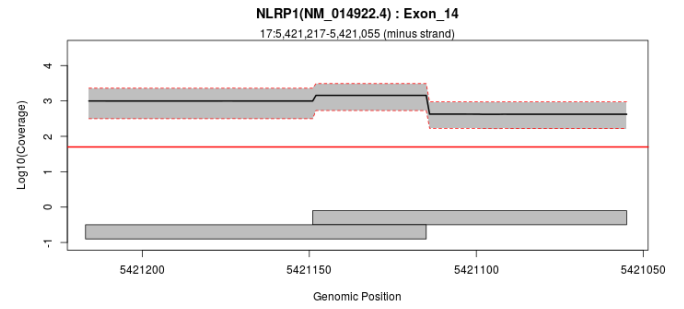
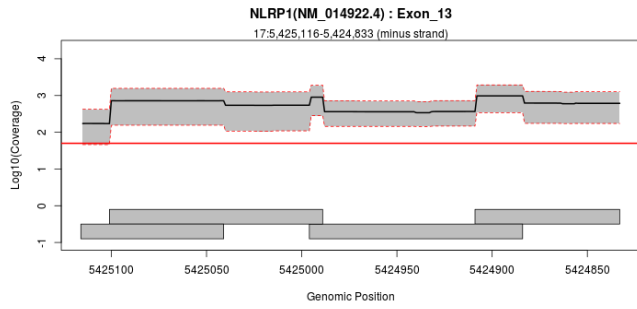




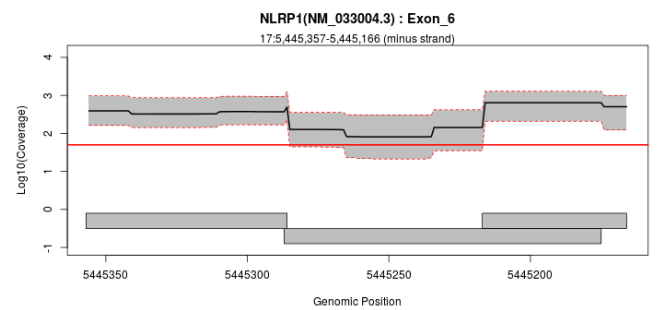
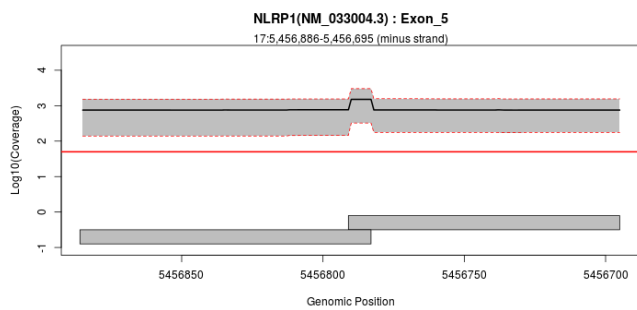
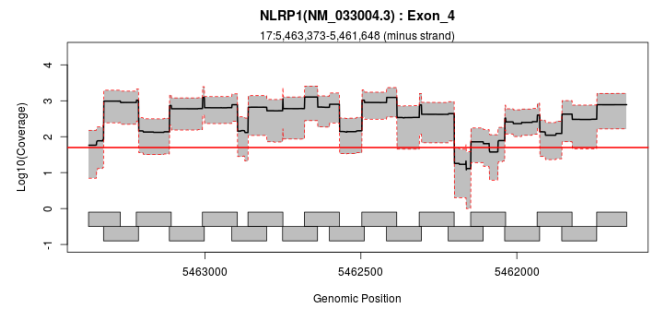
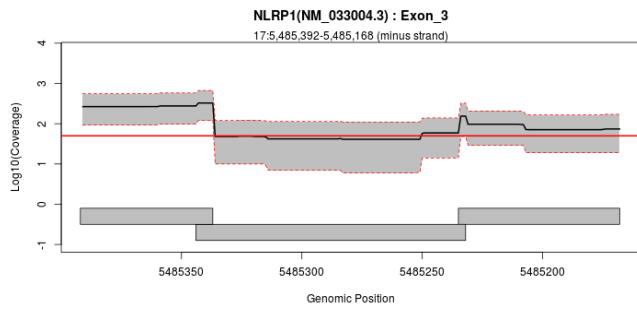
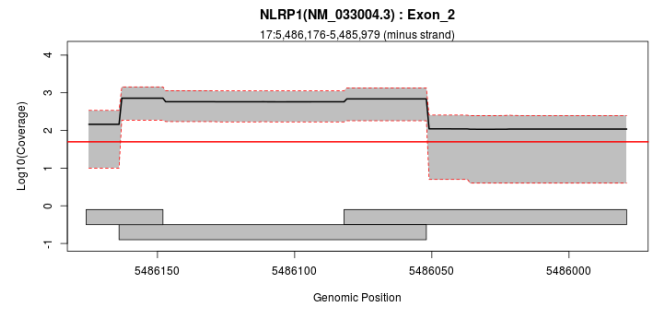
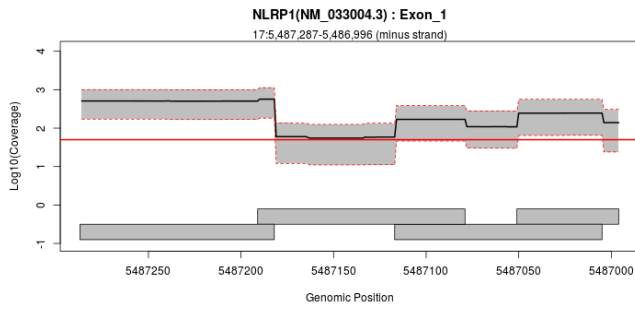
## Exon Overview: NLRP1 : NM\_014922.4



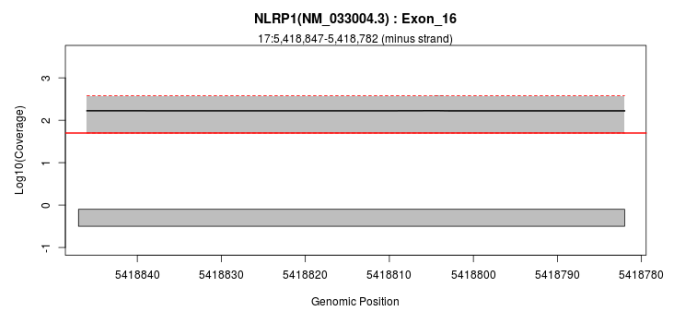
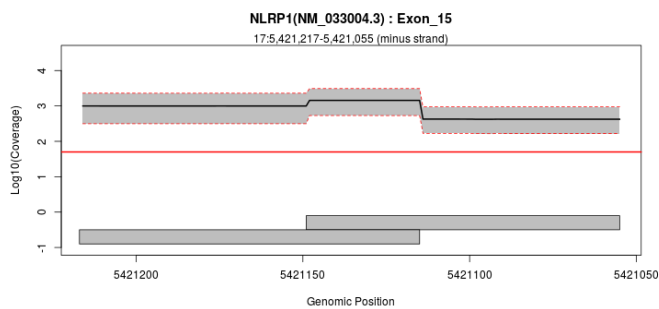
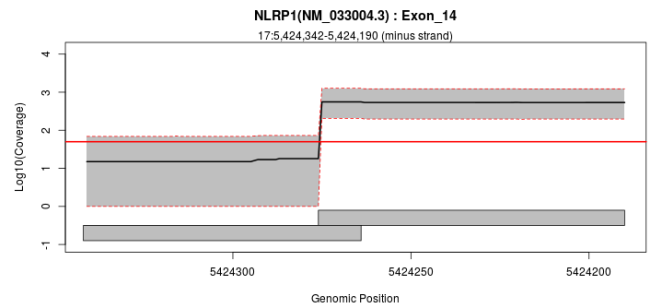
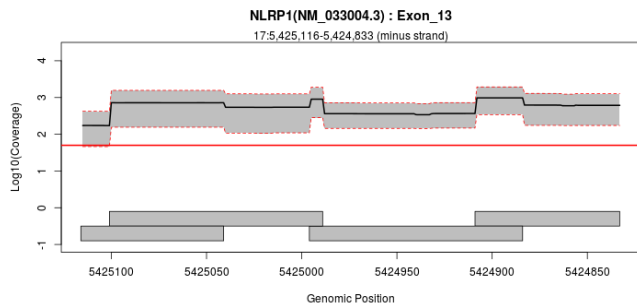
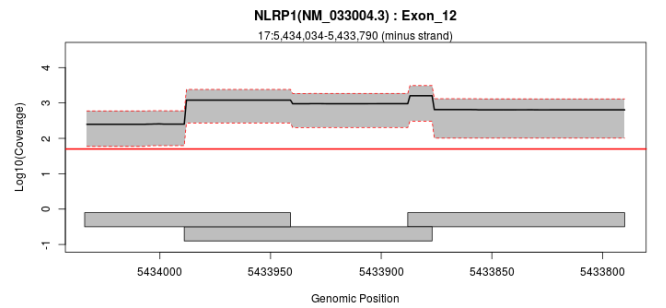
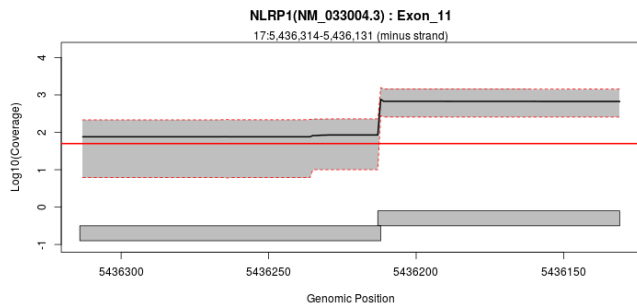
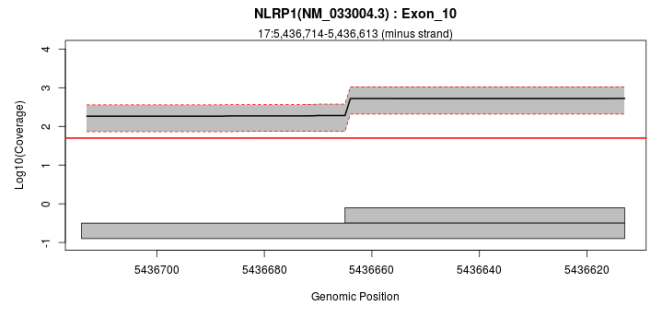
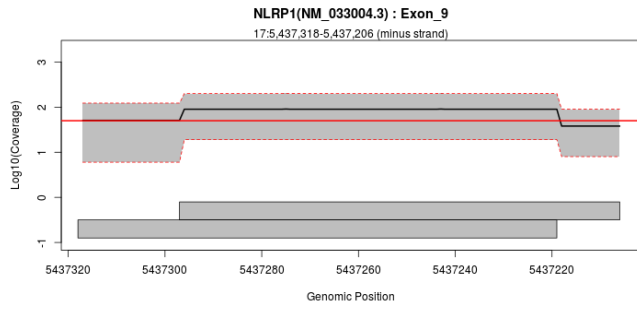
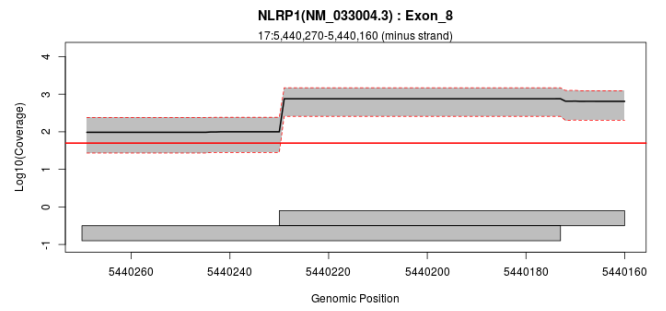
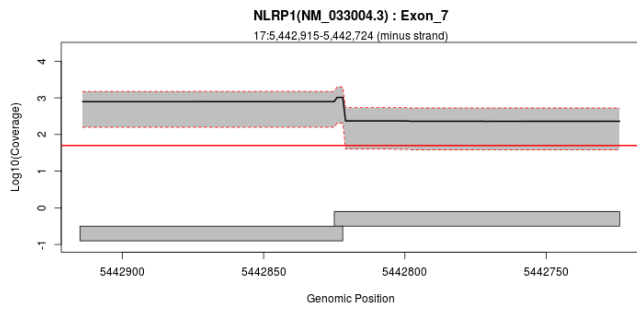


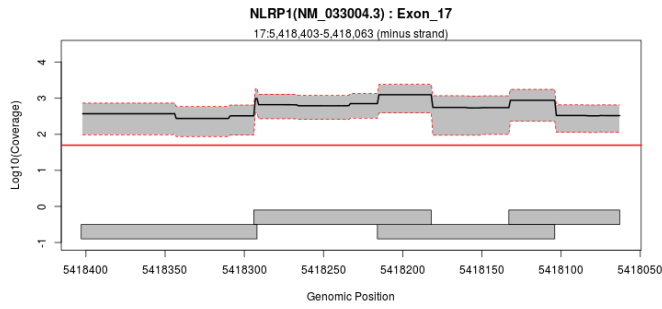


### Exon Overview: NLRP1 : NM\_033004.3

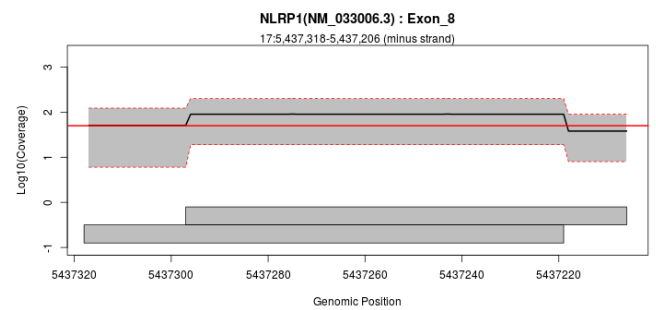
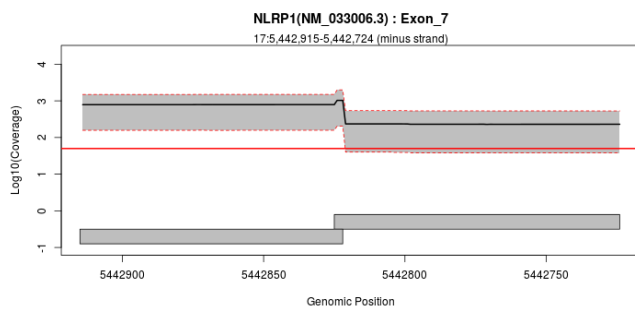
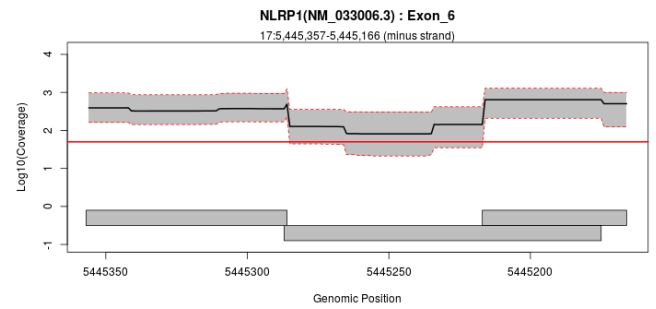
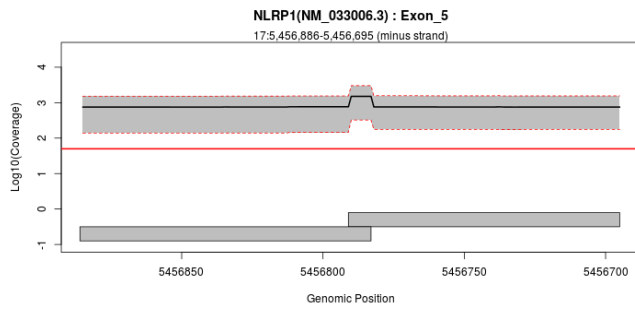
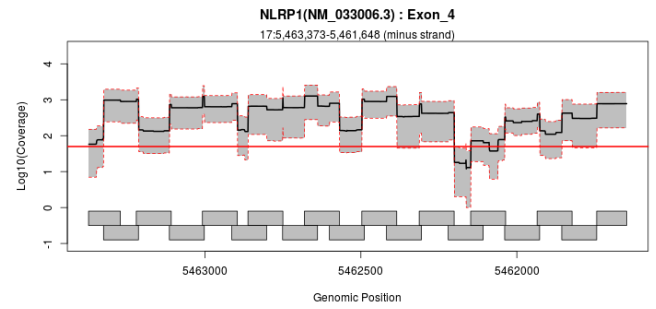
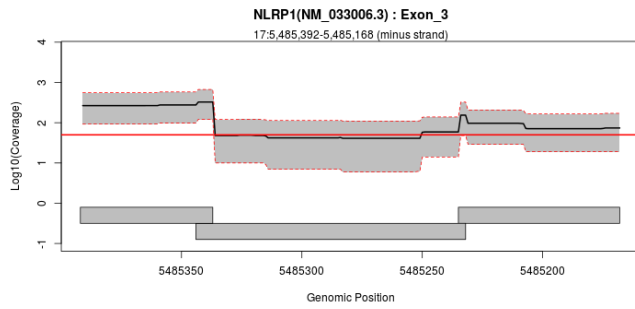
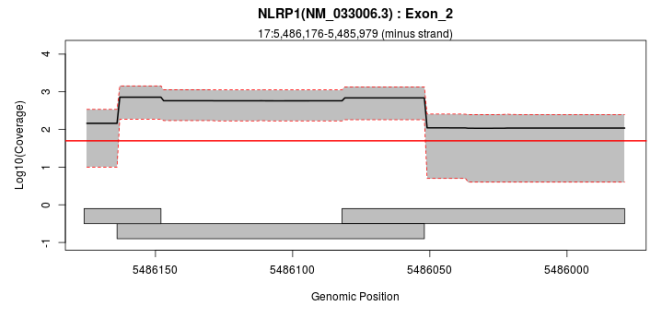
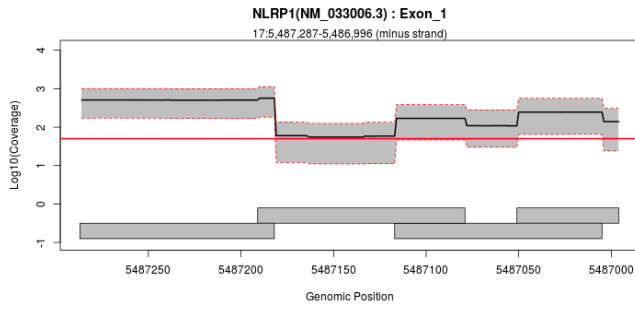


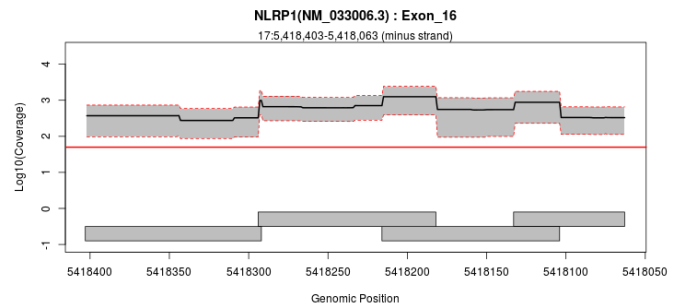
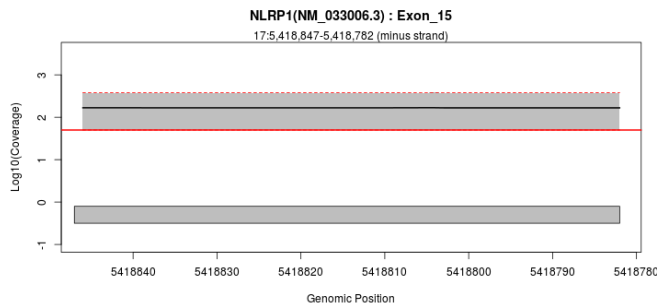
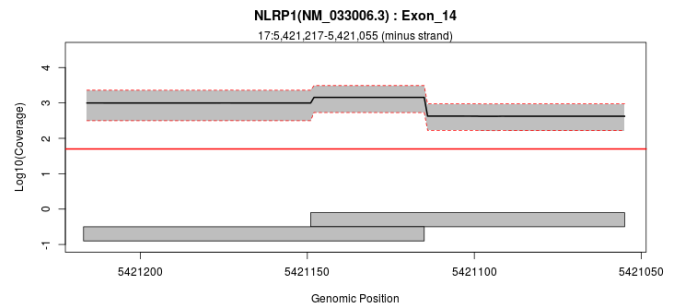
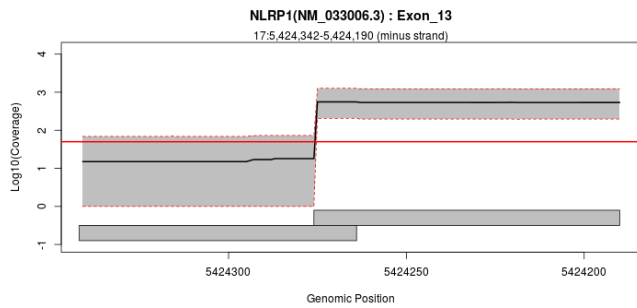
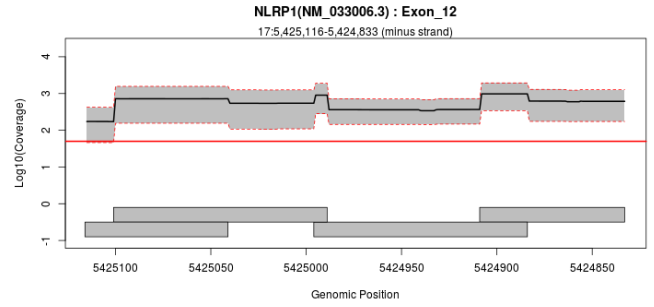
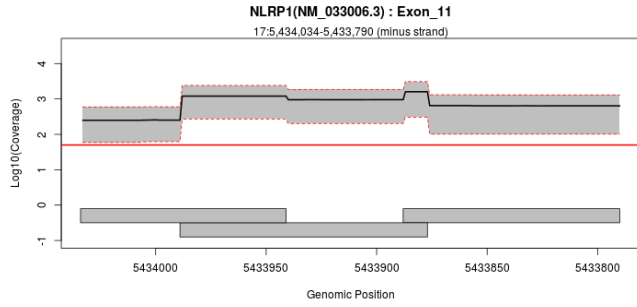
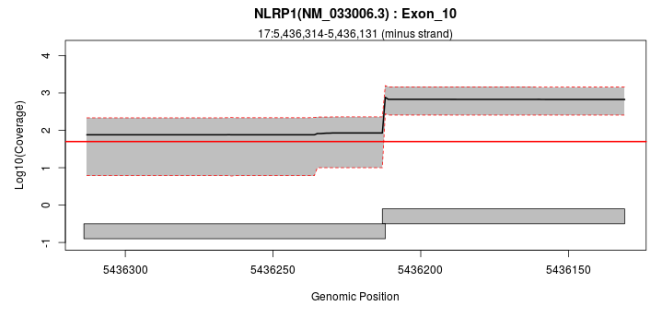
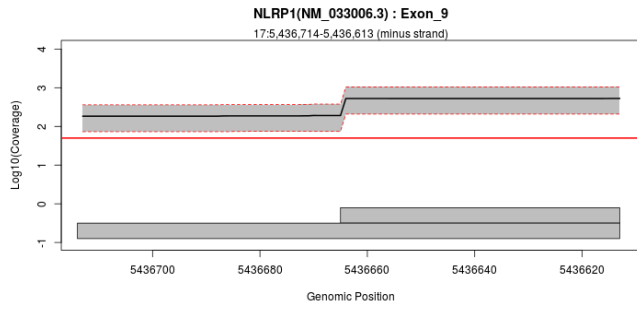




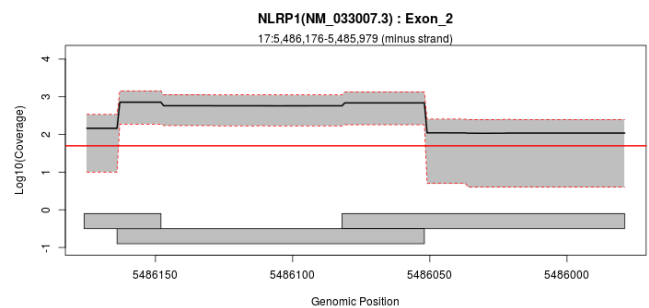
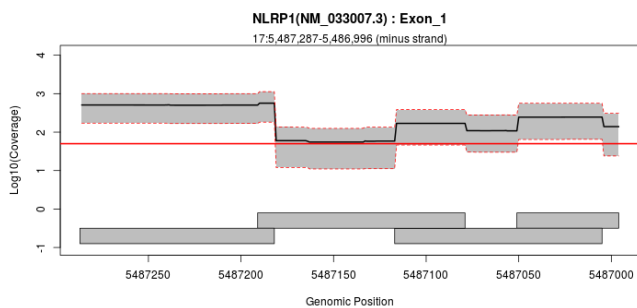


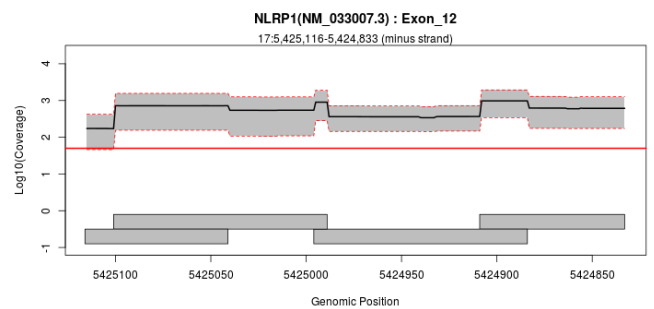
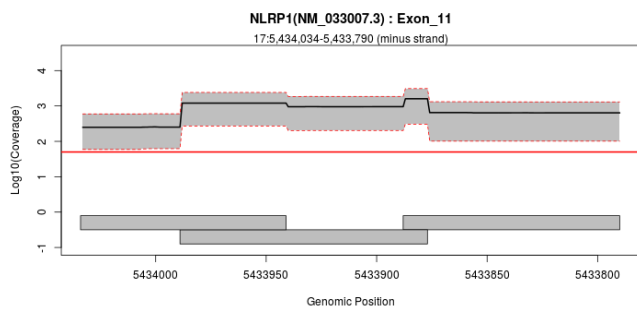
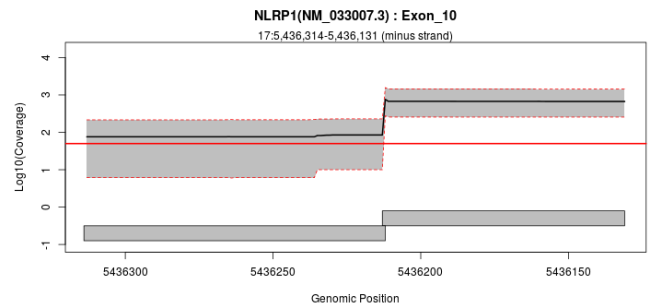
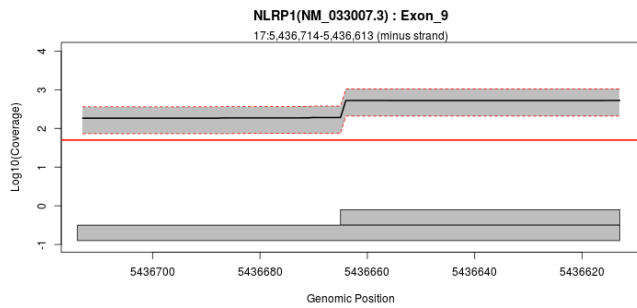
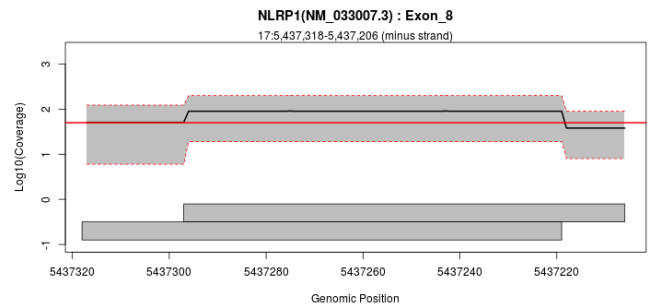
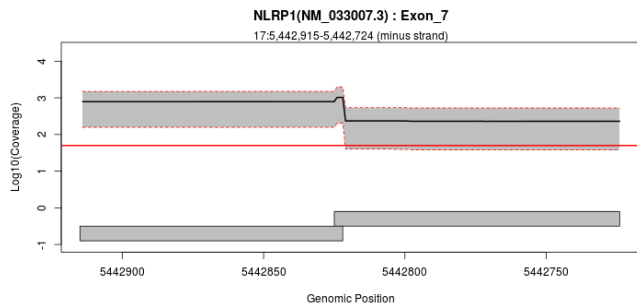
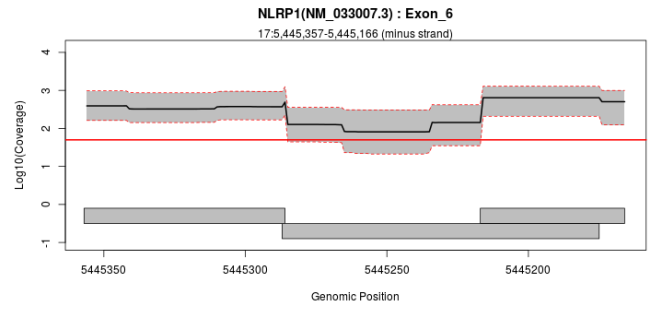
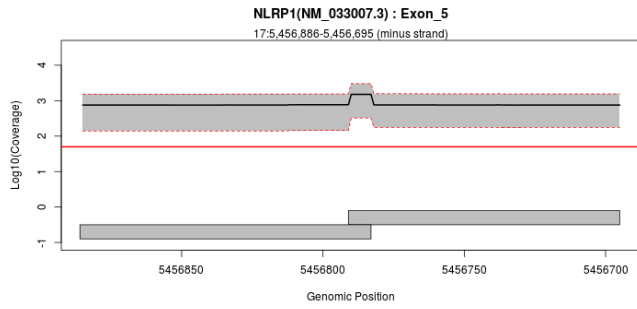
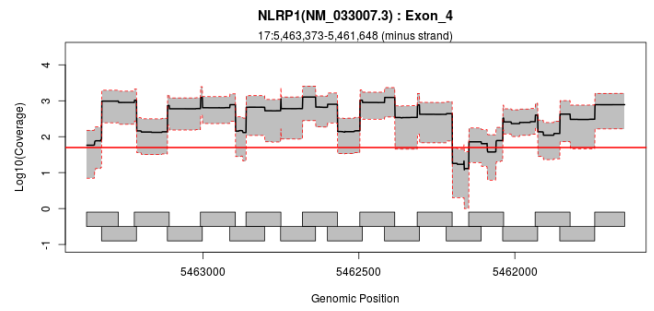
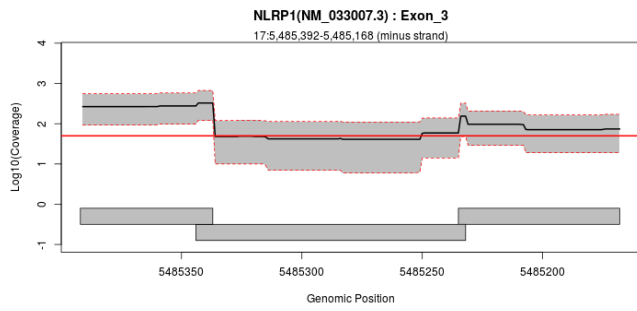
## Exon Overview: NLRP1 : NM\_033006.3

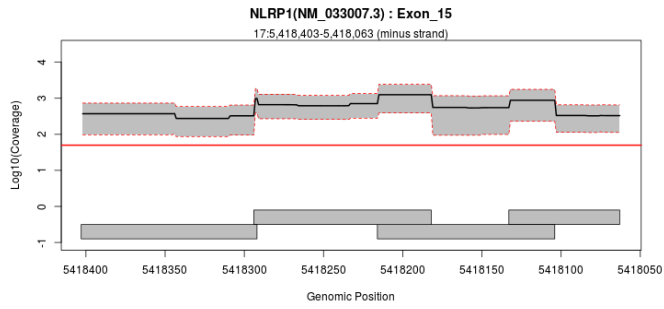
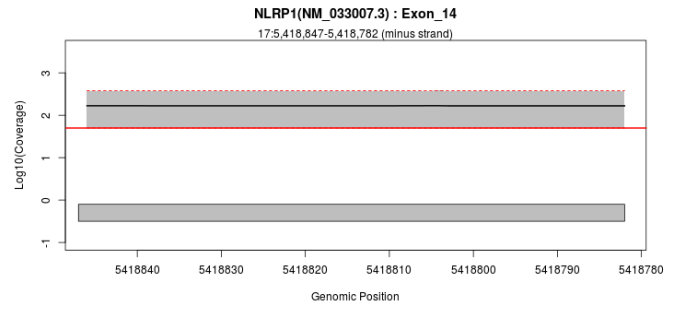
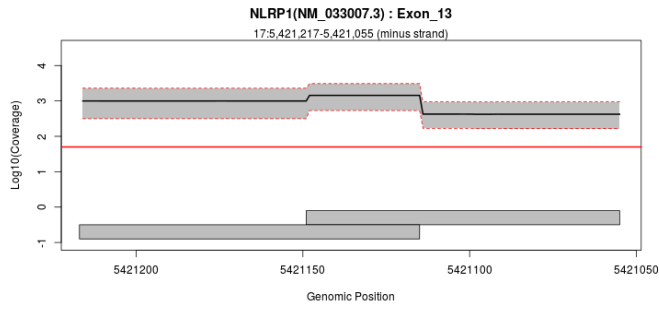




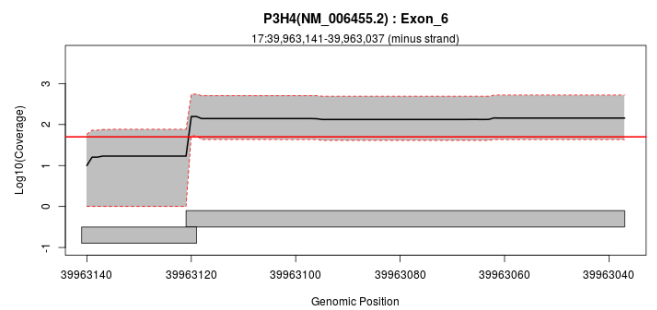
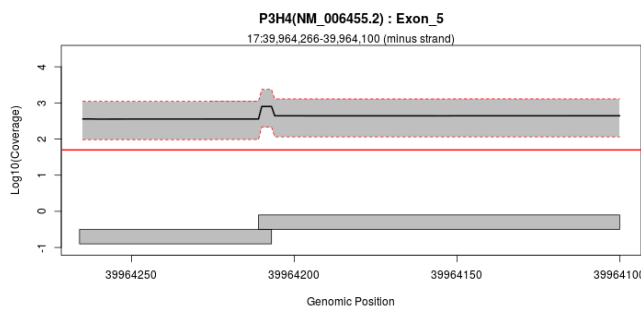
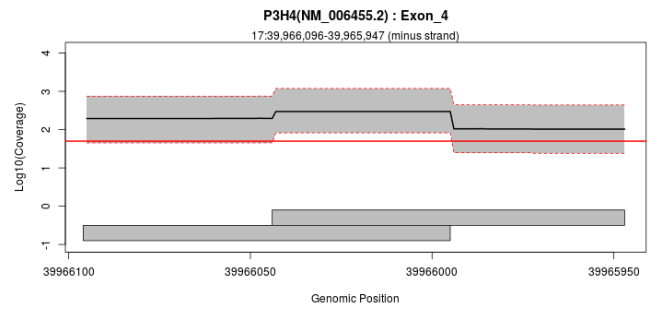
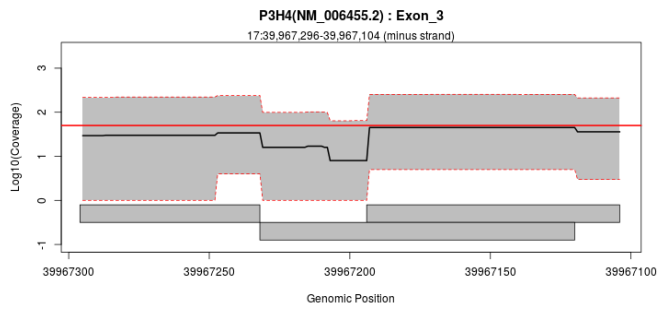
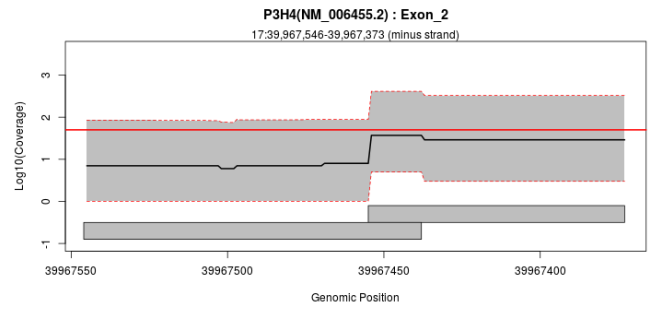
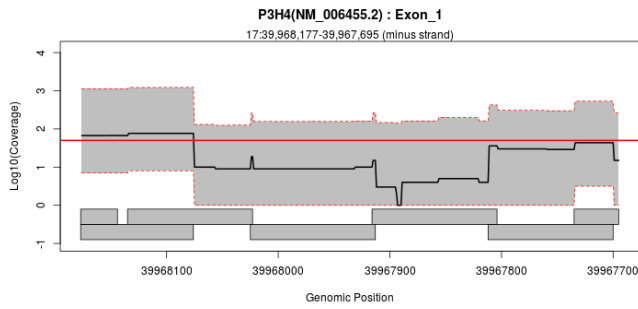
## Exon Overview: NLRP1 : NM\_033007.3

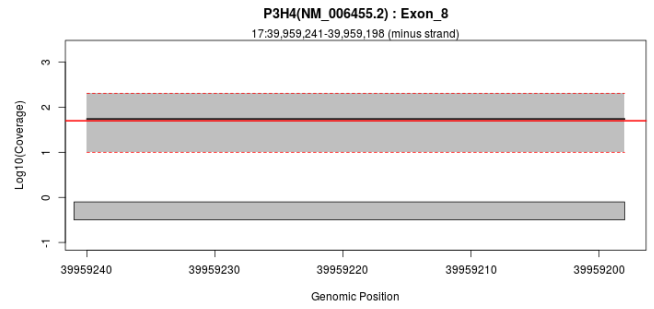
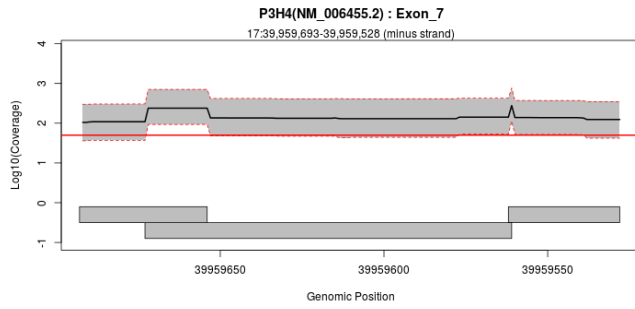




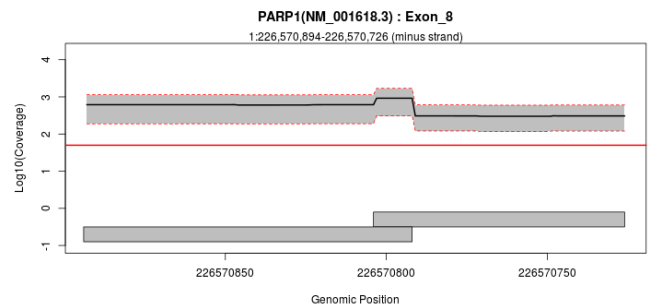
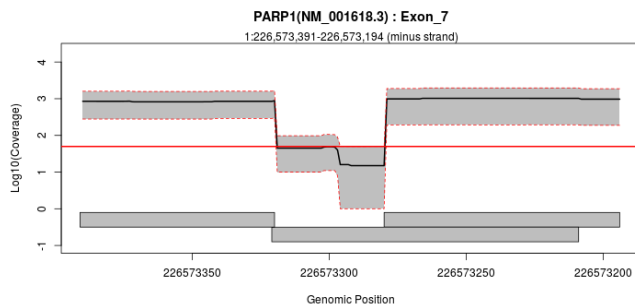
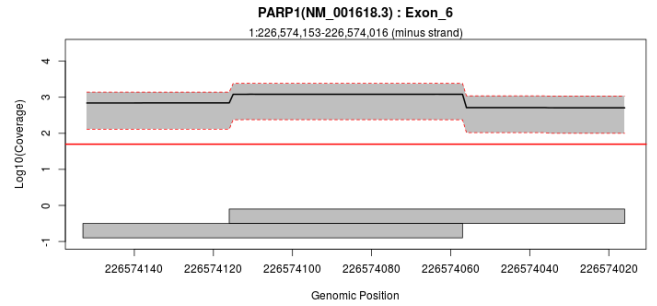
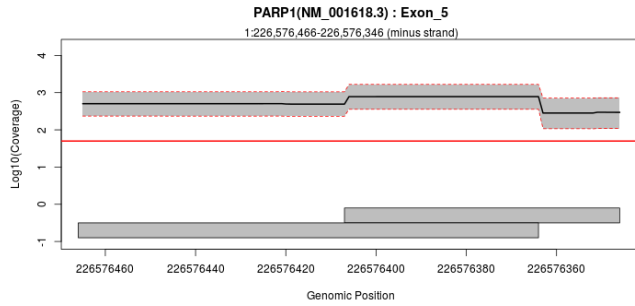
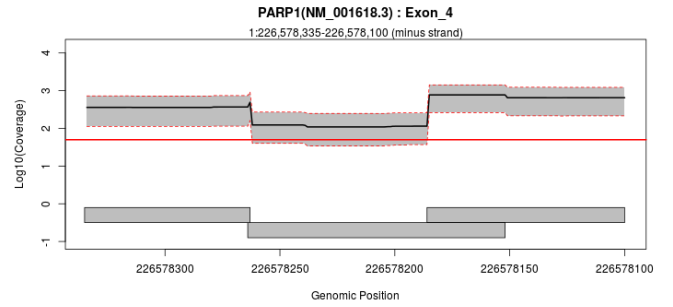
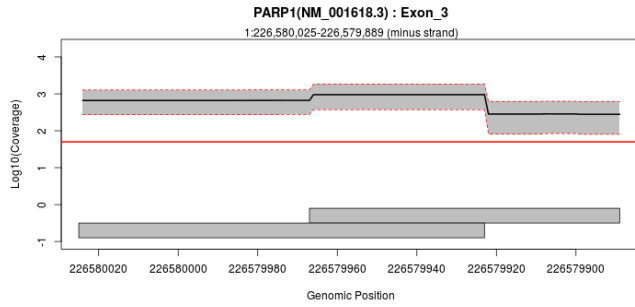
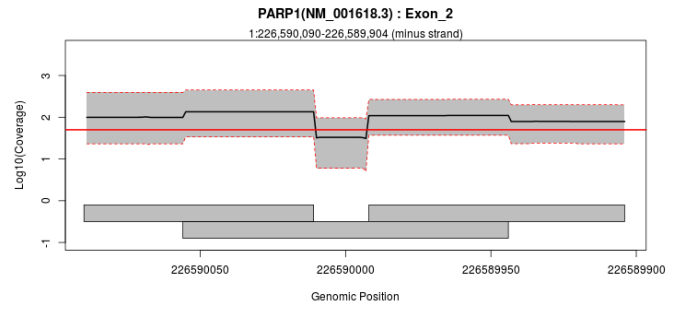
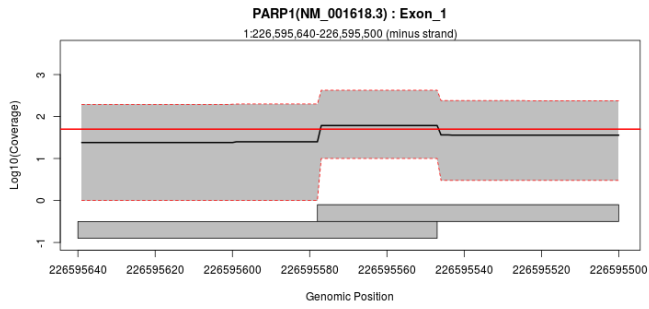


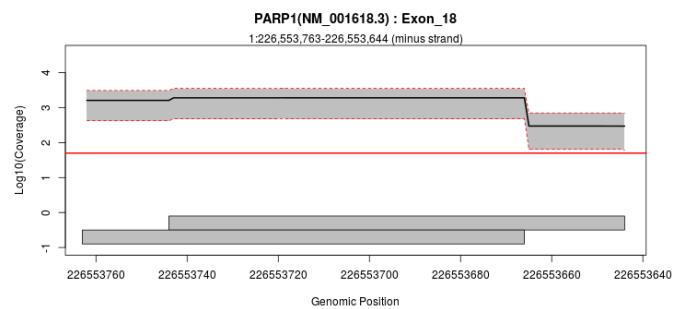
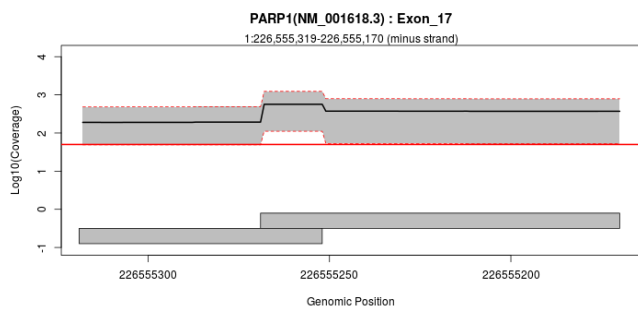
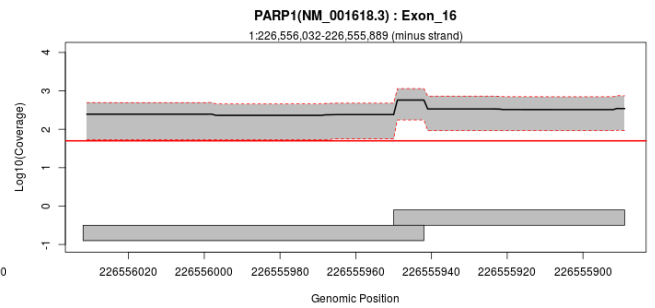
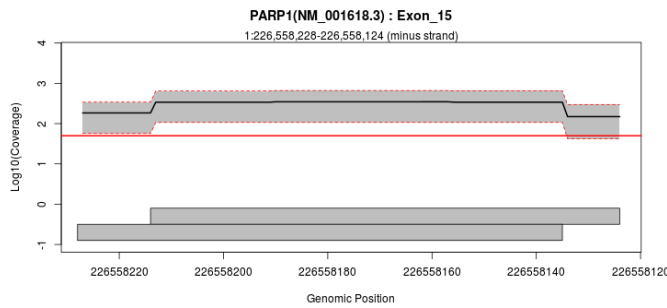
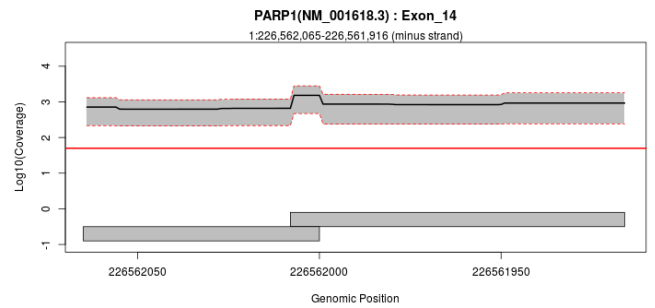
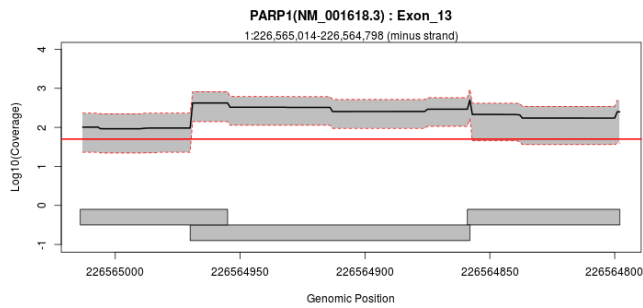
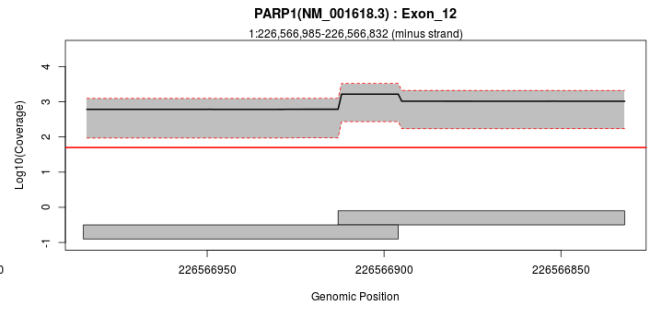
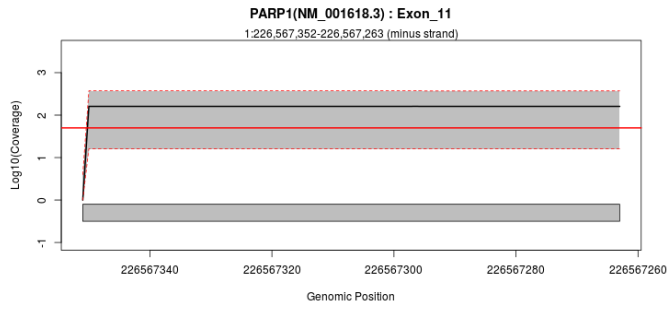
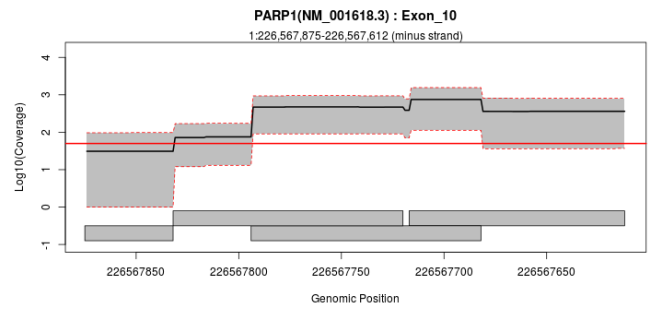
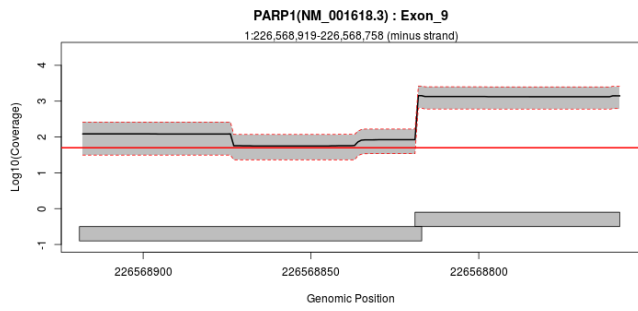
## Exon Overview: P3H4 : NM\_006455.2

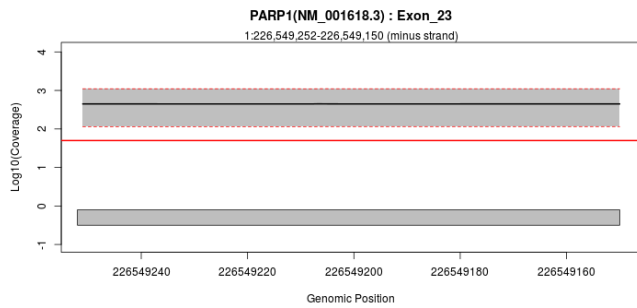
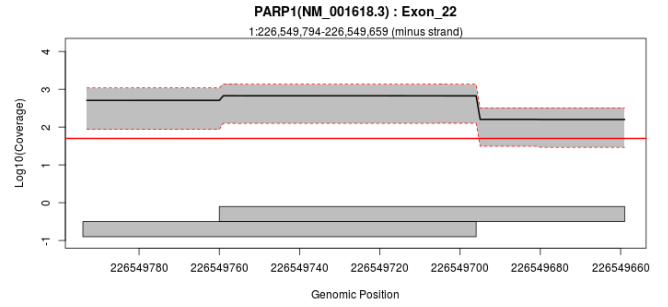
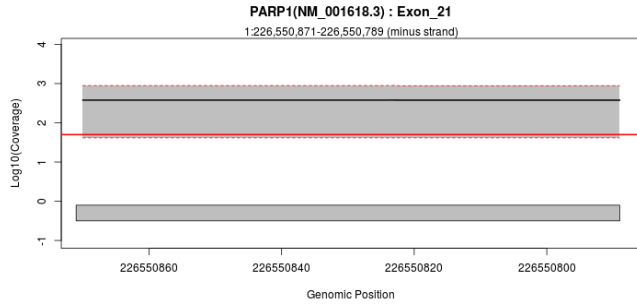
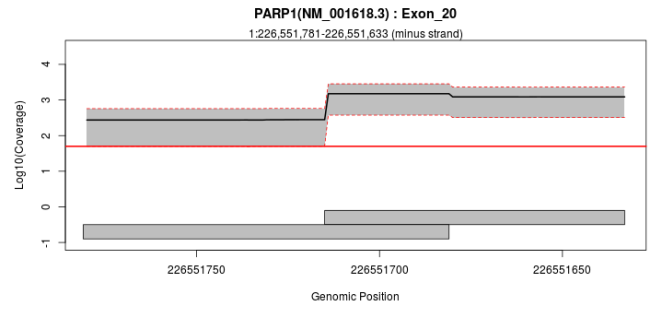
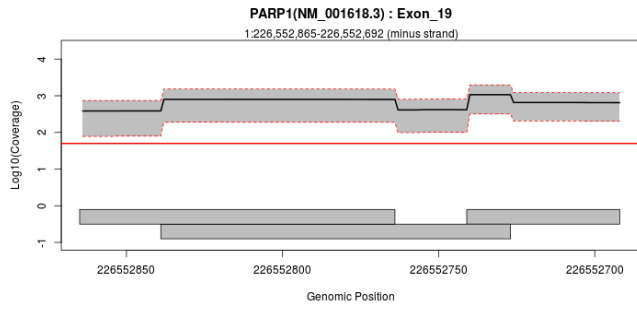




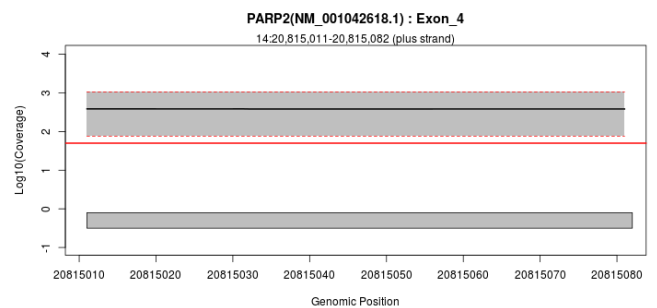
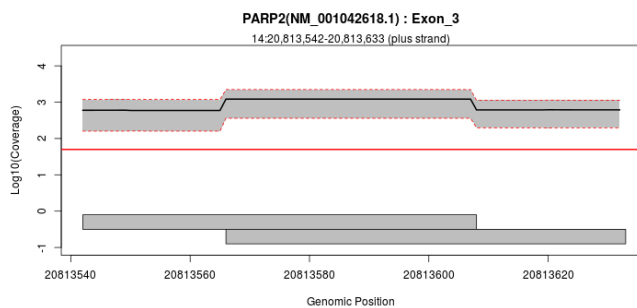
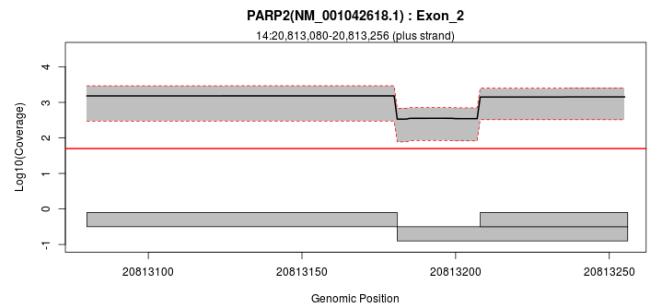
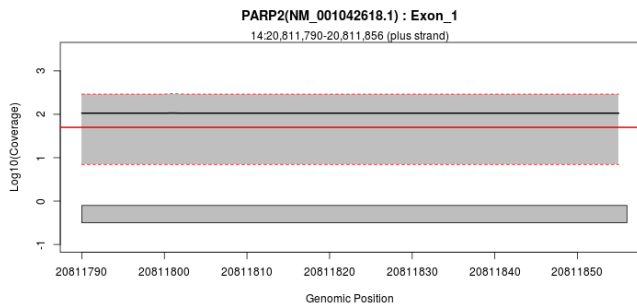
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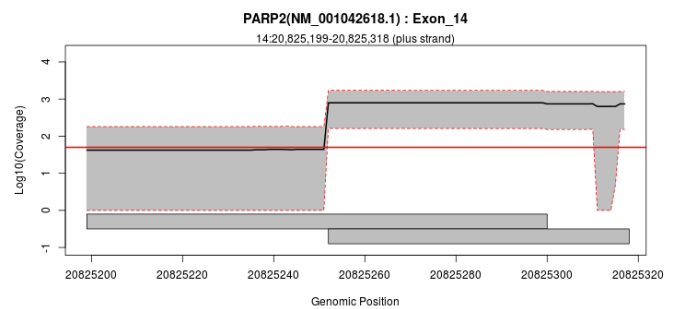
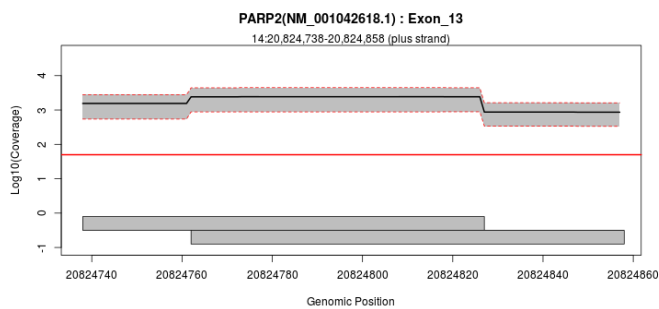
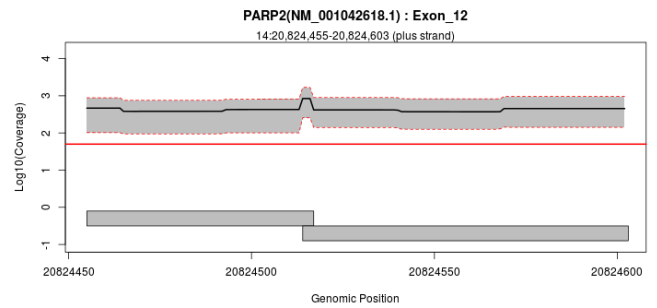
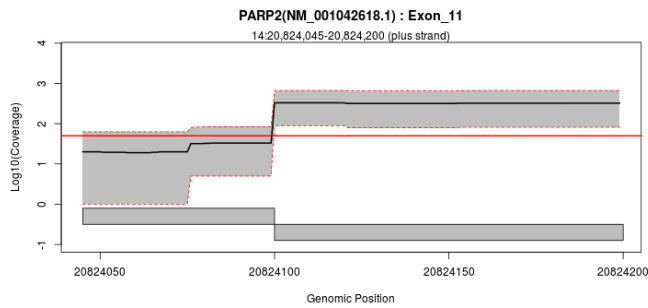
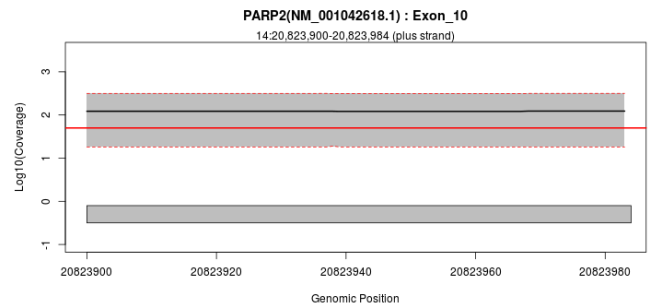
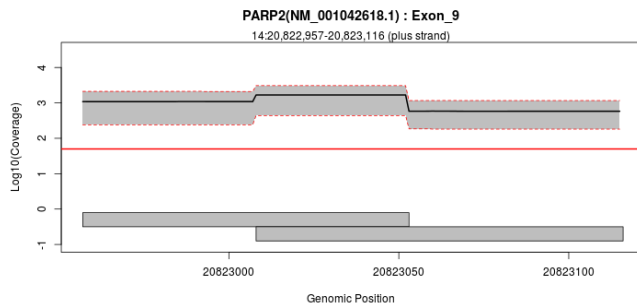
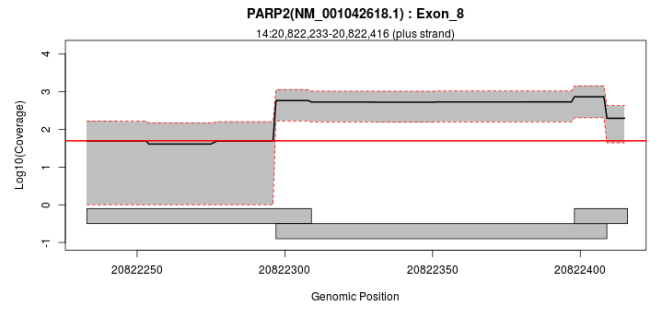
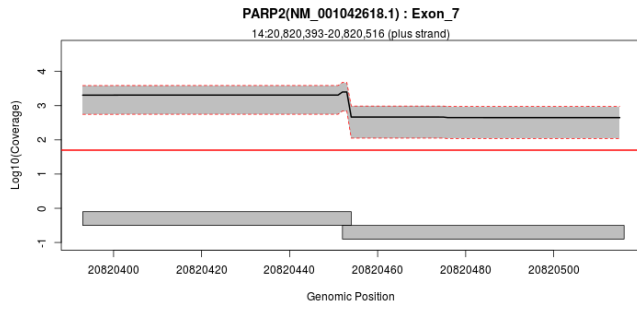
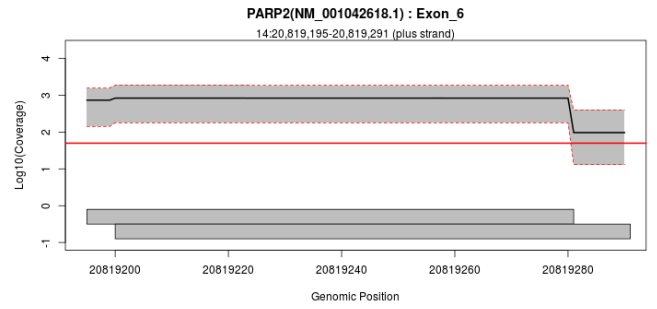
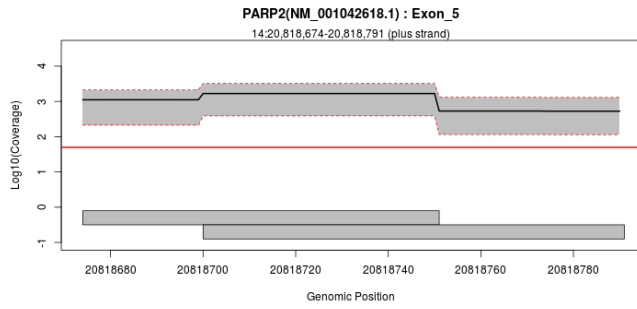


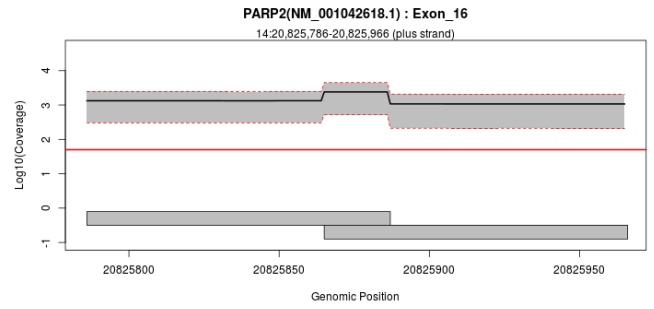
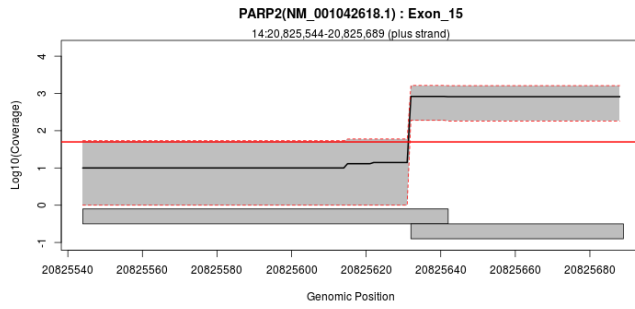


## Exon Overview: PARP2 : NM\_001042618.1

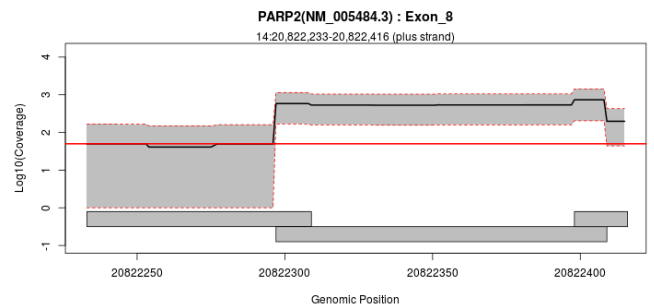
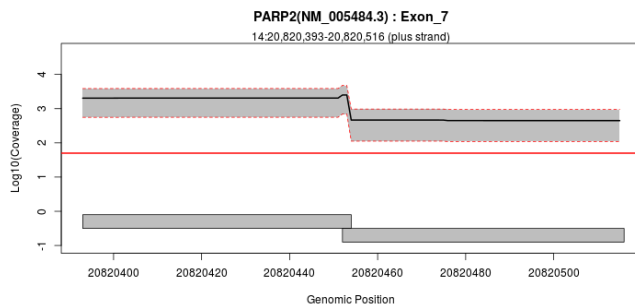
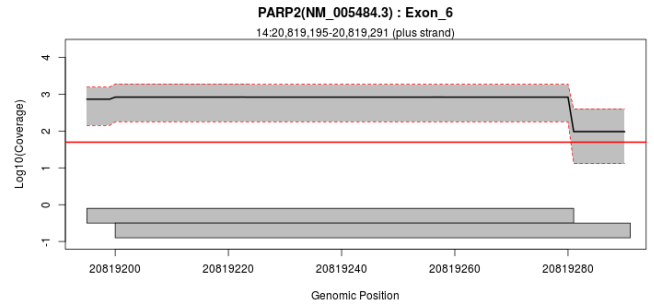
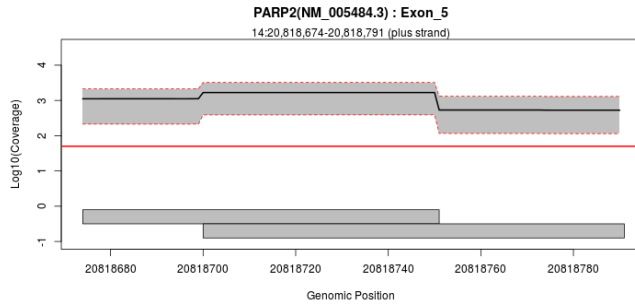
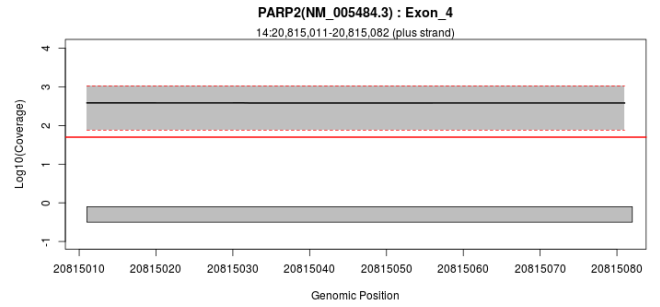
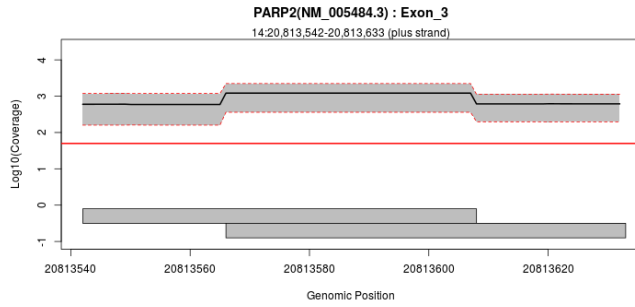
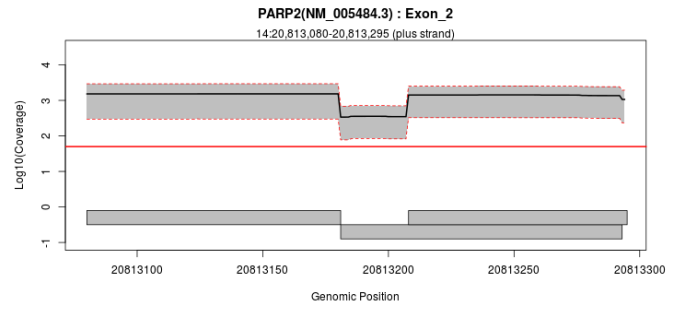
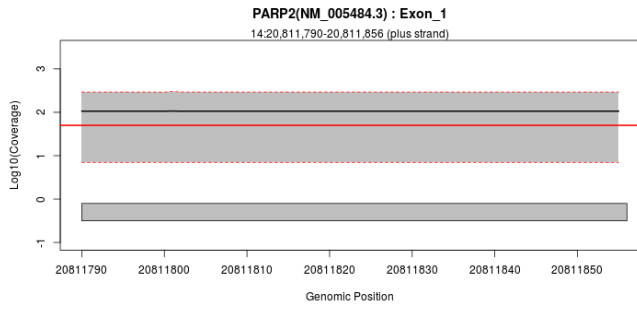


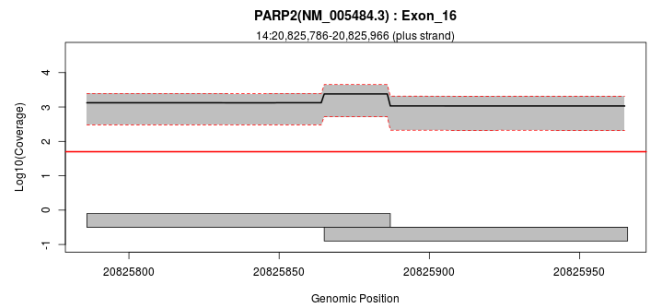
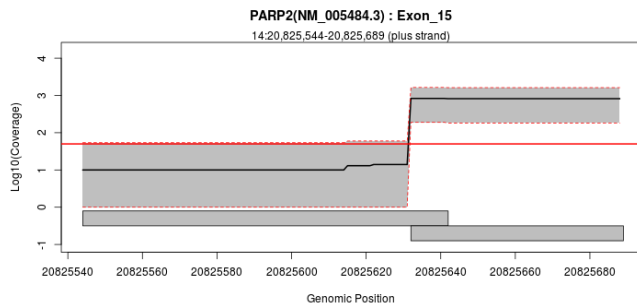
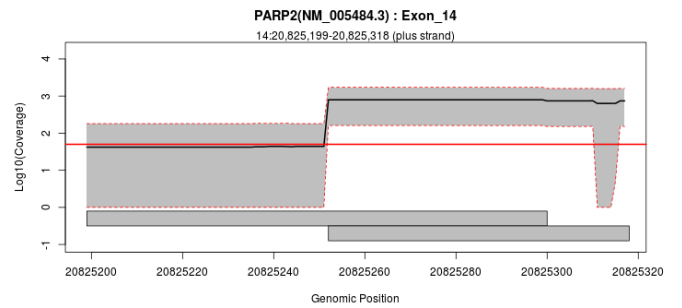
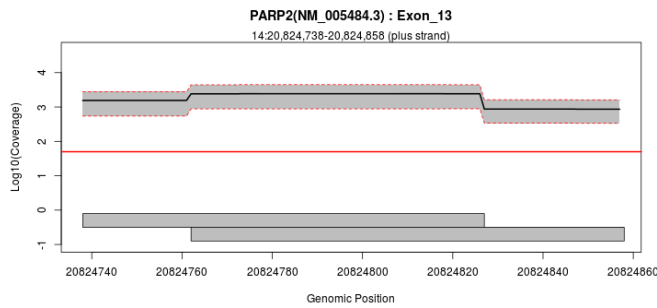
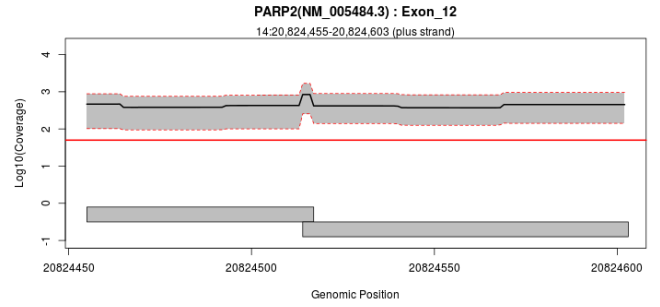
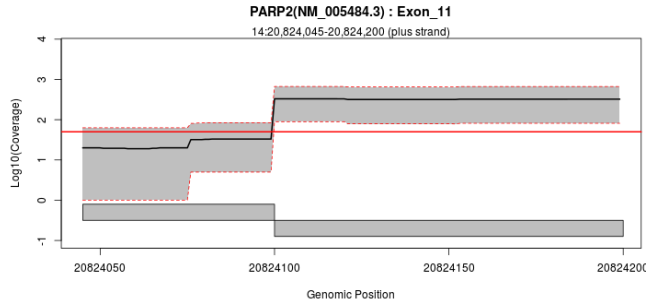
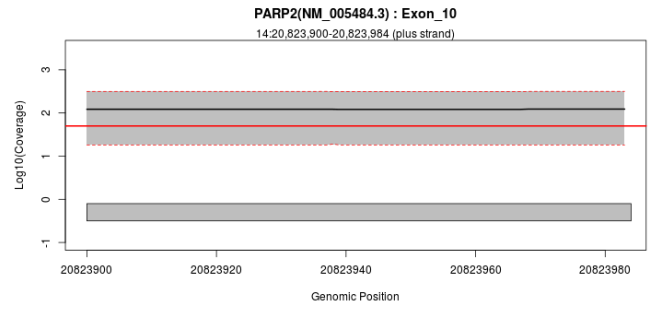
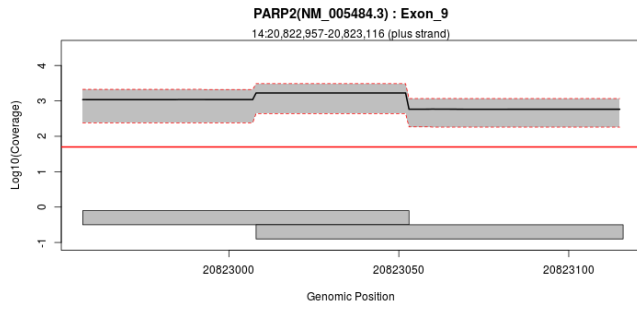




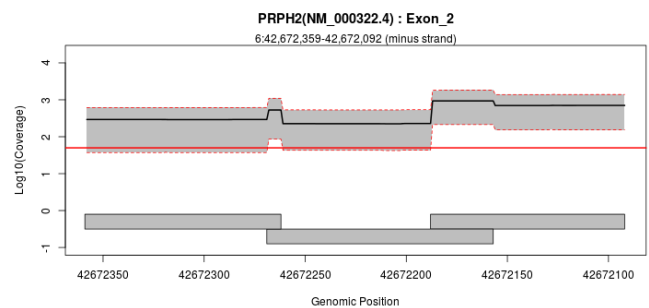
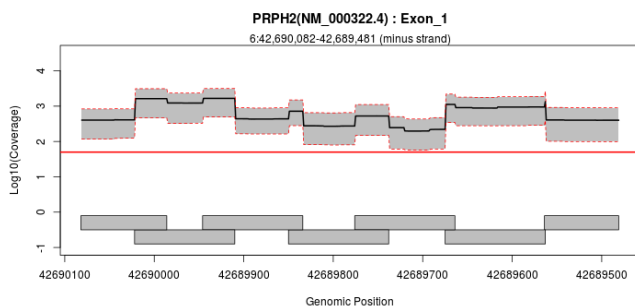


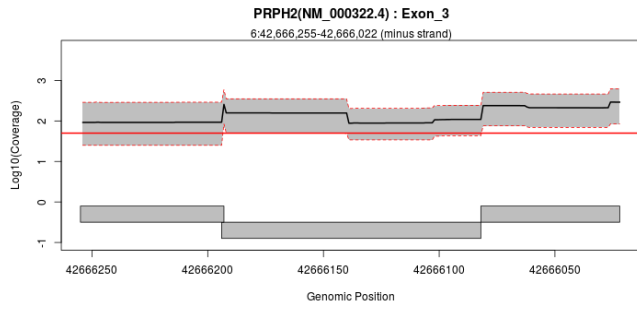
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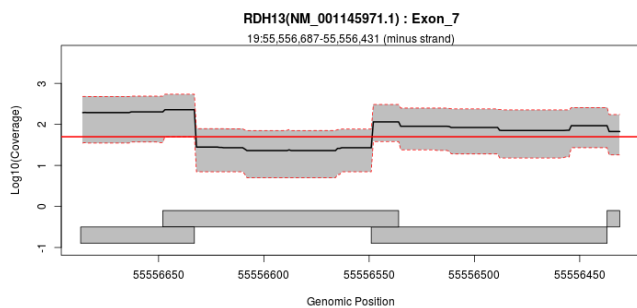
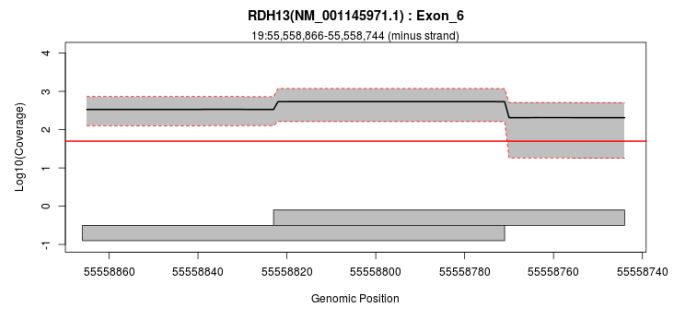
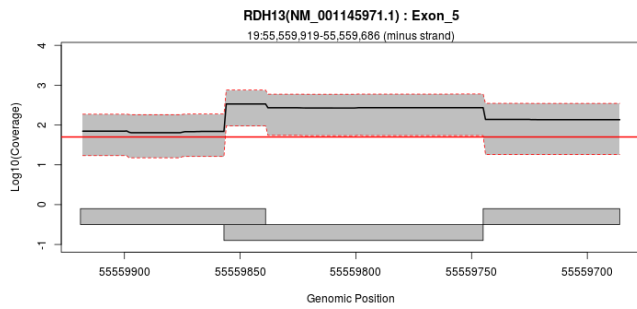
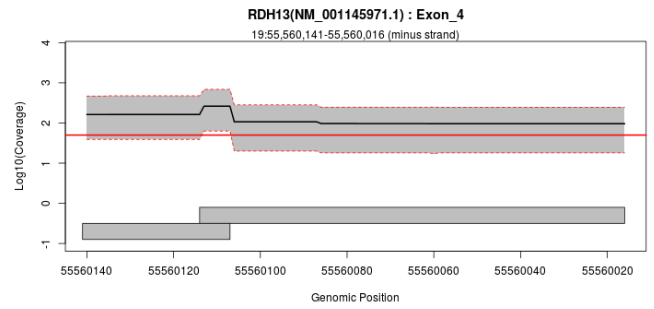
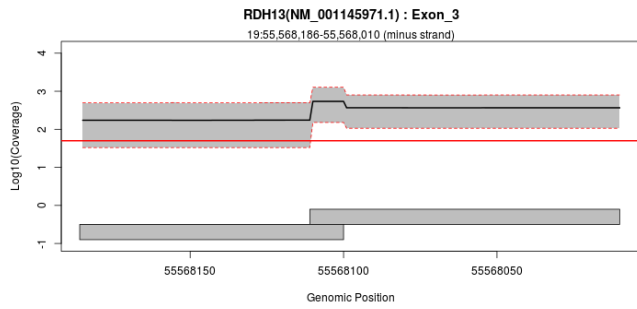
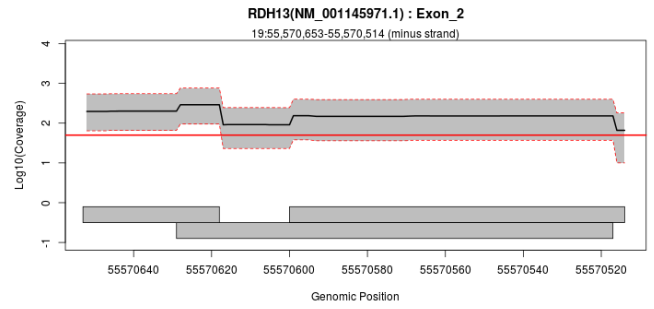
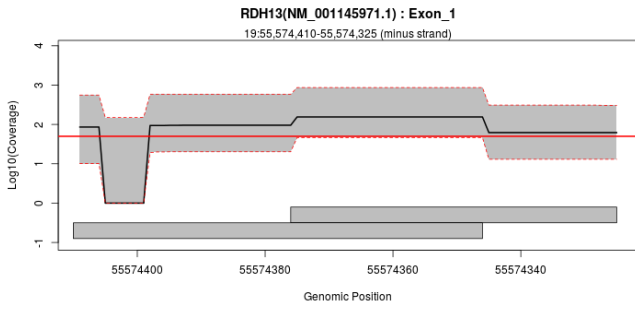


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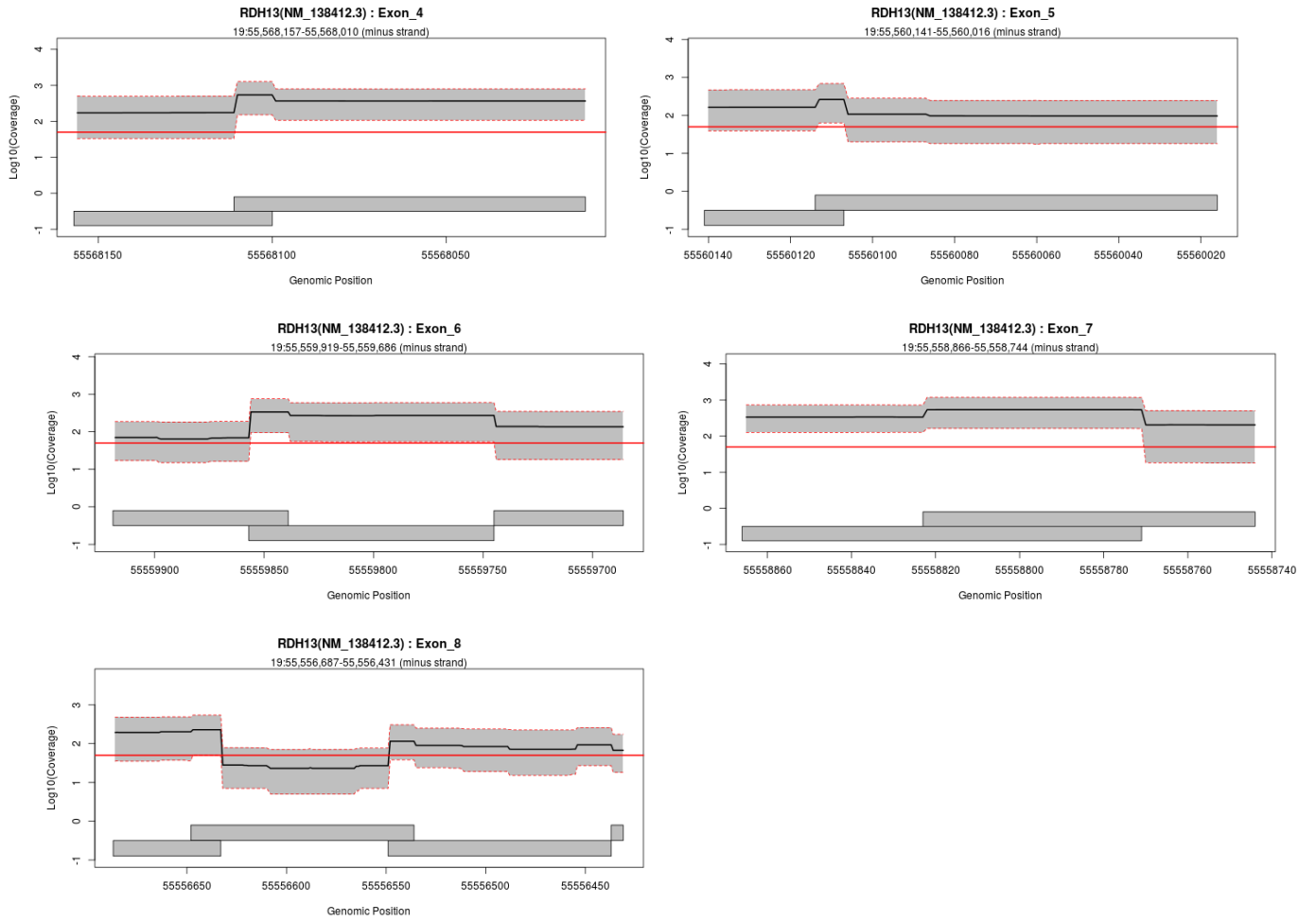




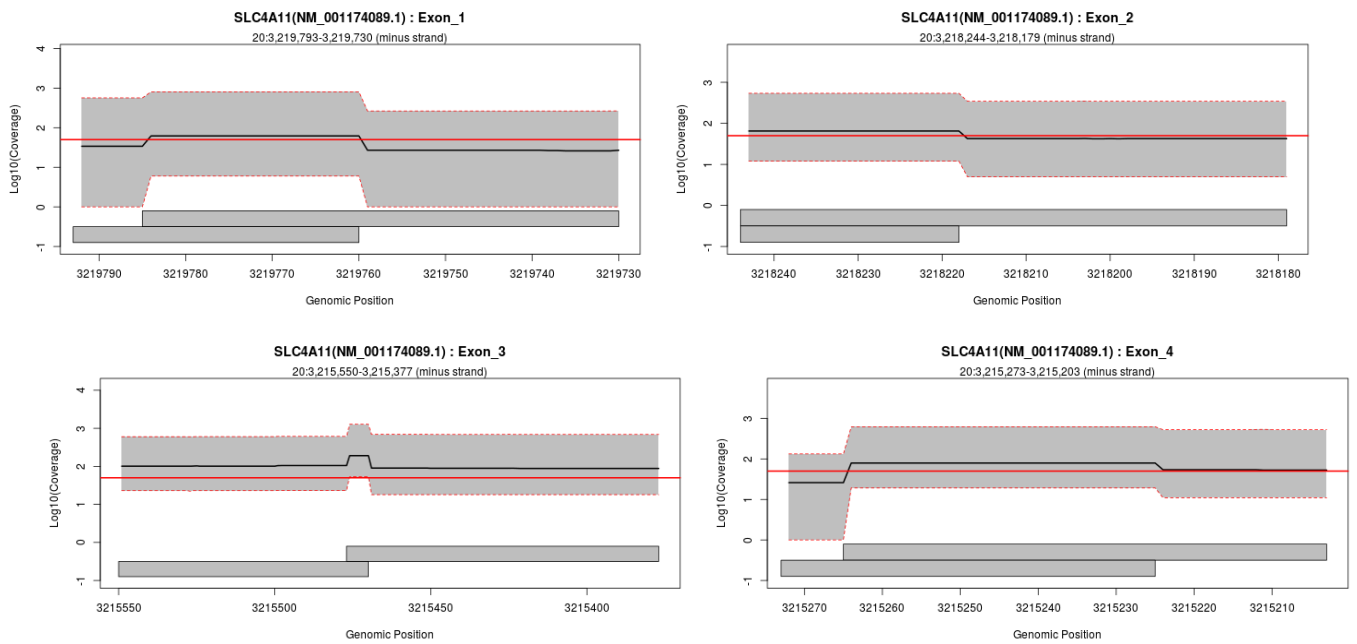
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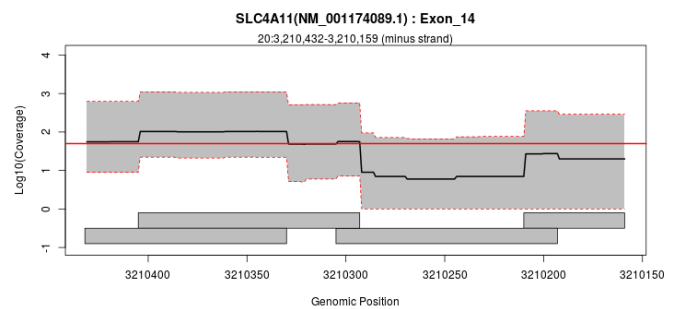
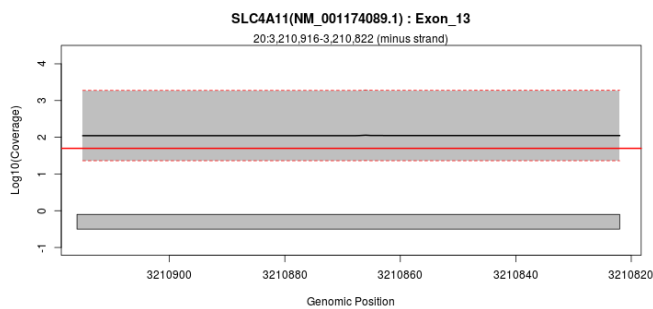
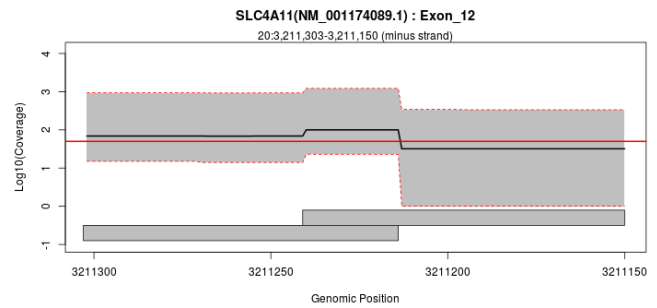
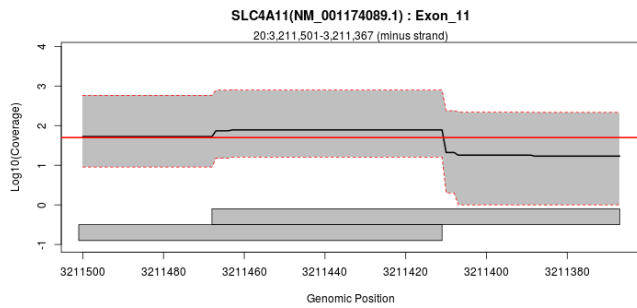
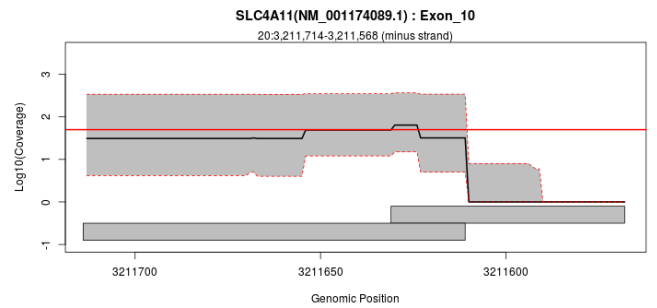
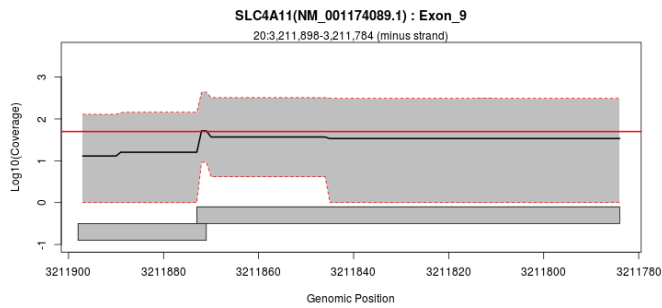
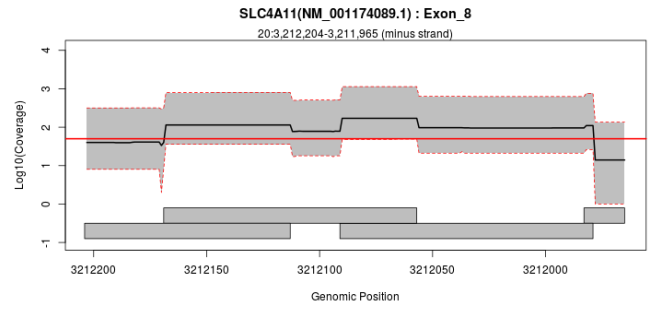
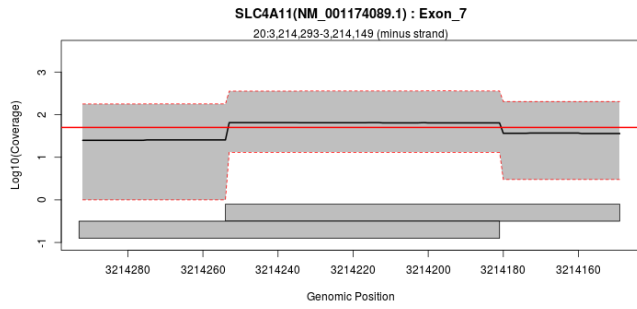
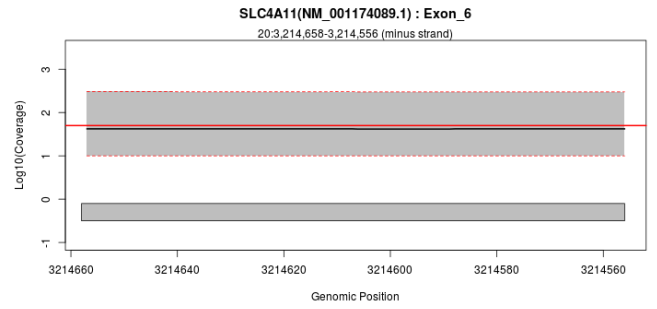
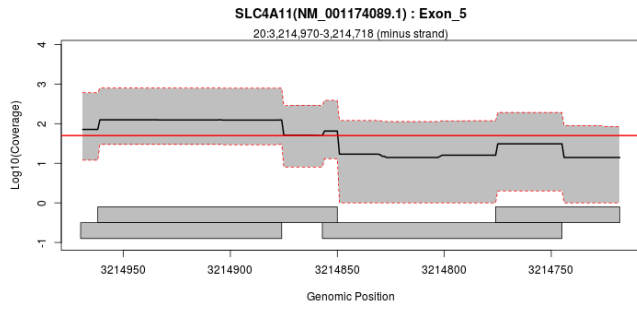


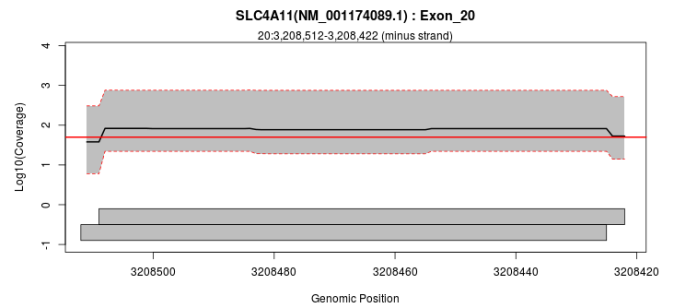
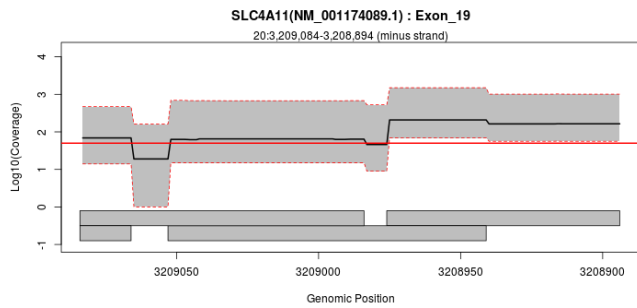
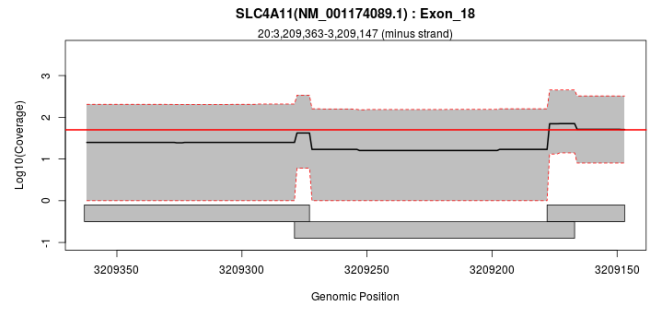
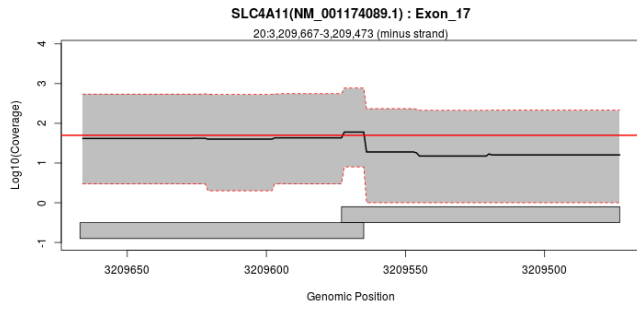
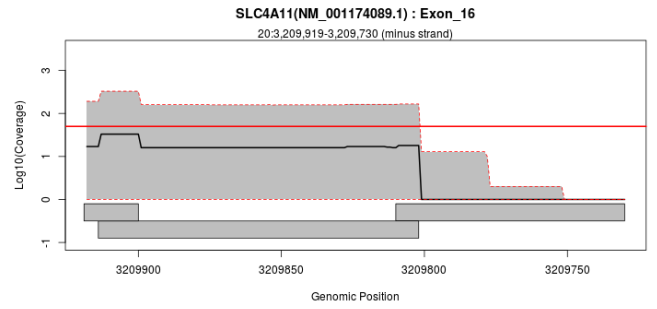
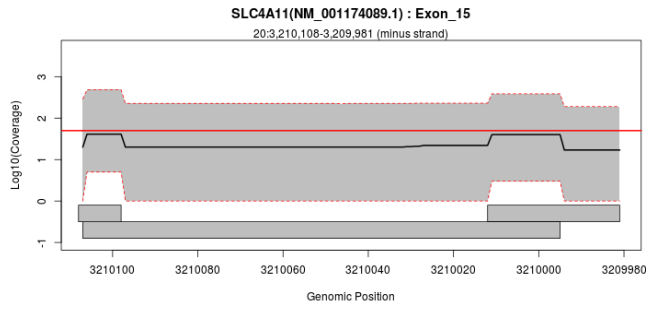
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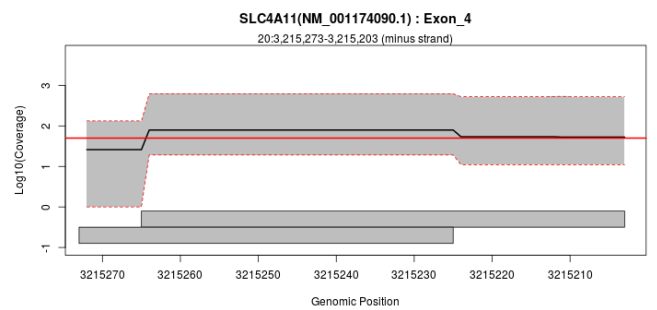
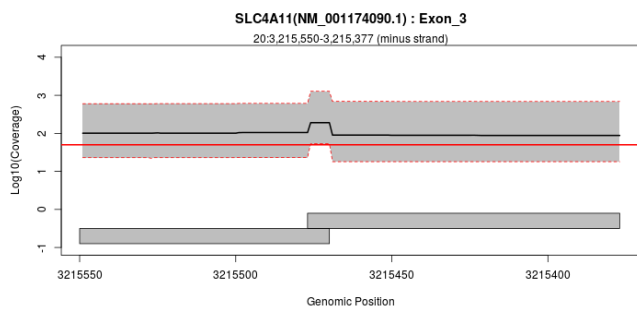
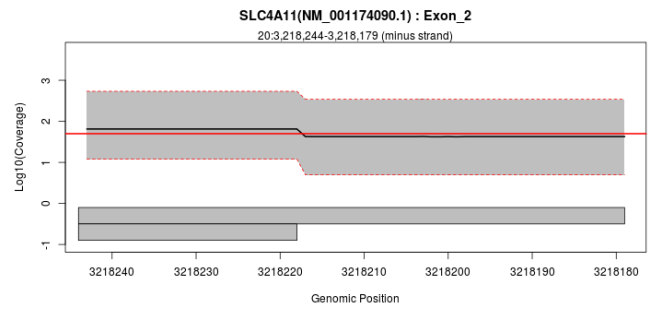
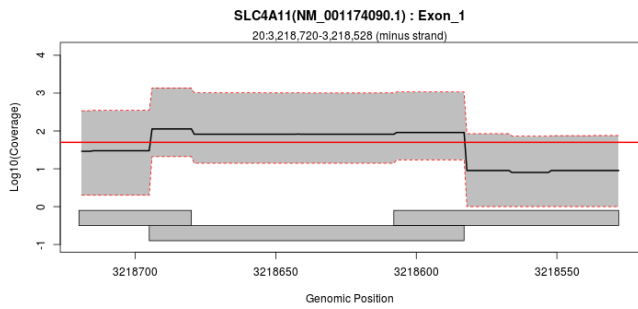
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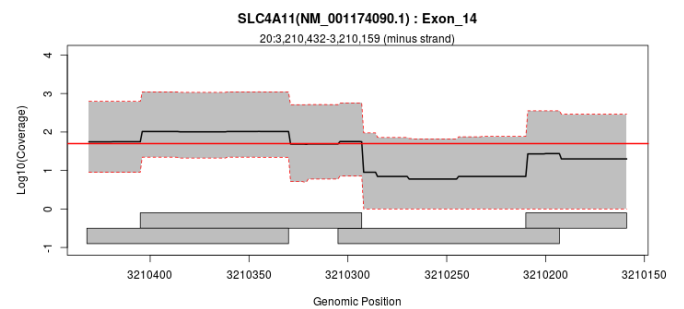
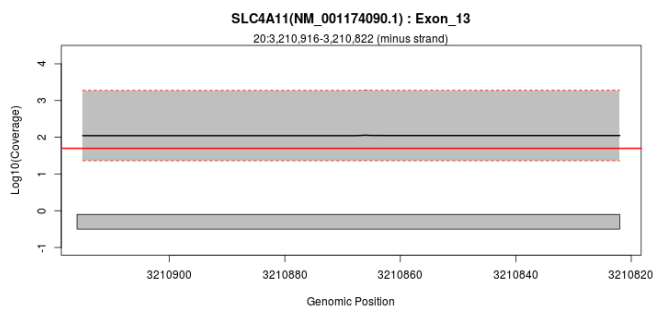
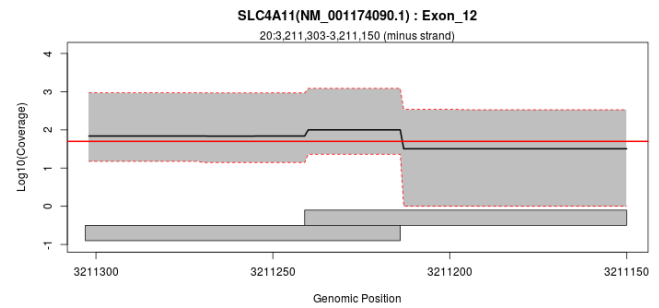
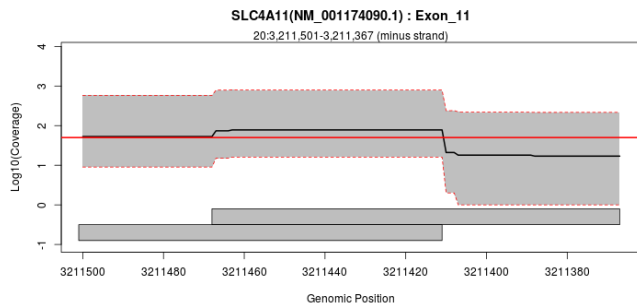
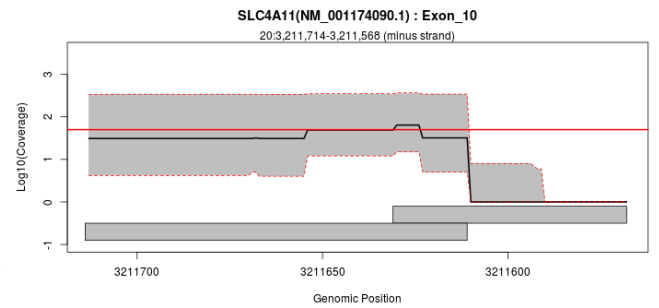
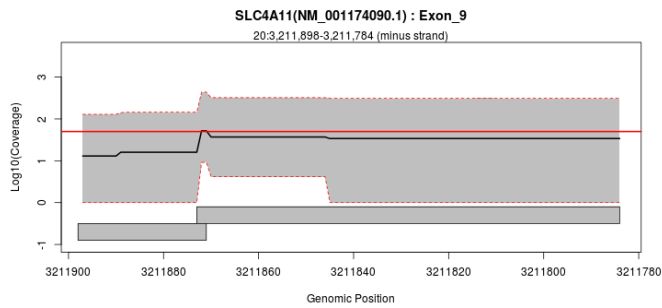
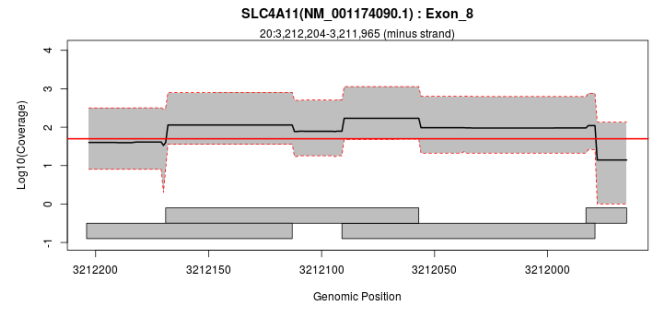
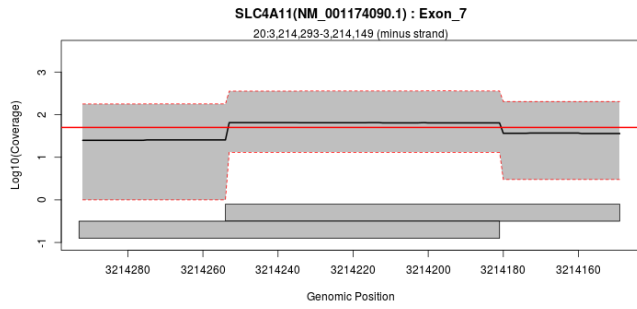
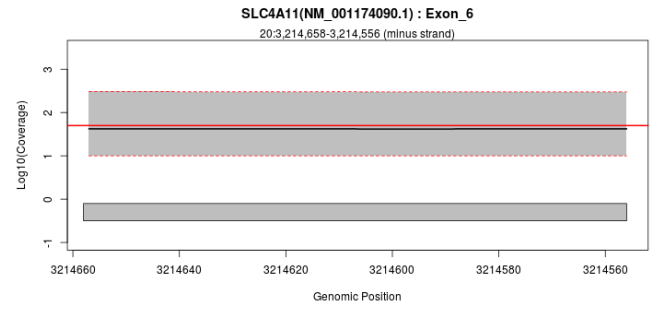
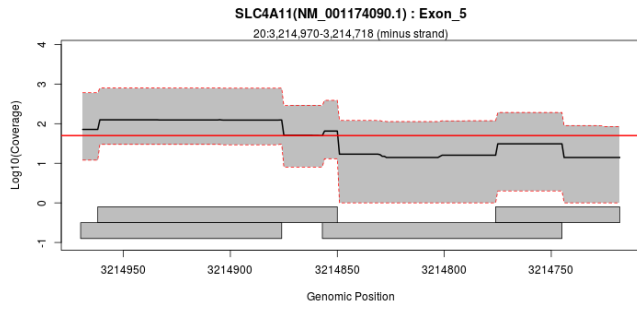




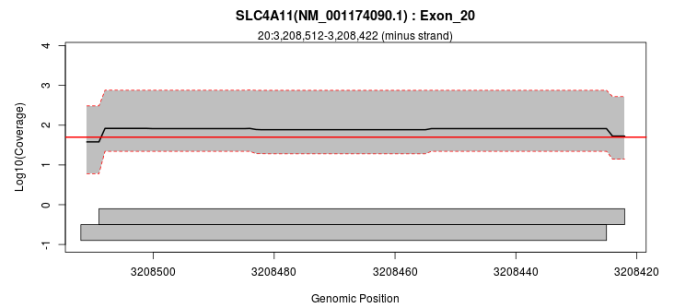
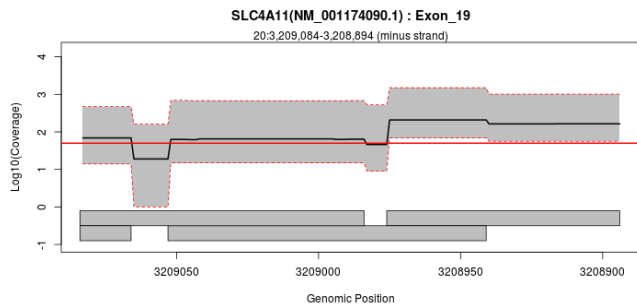
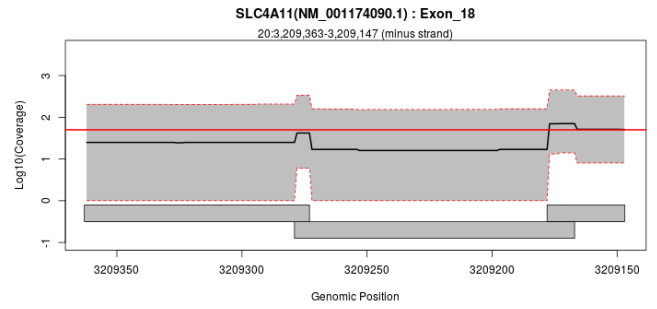
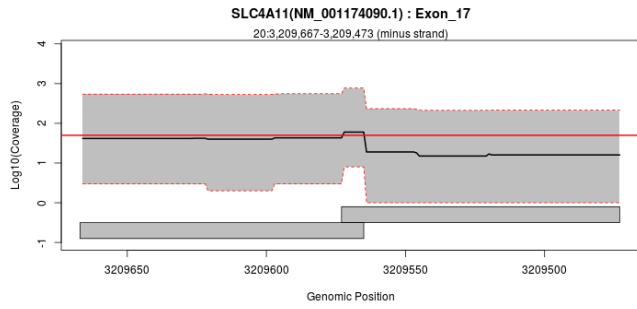
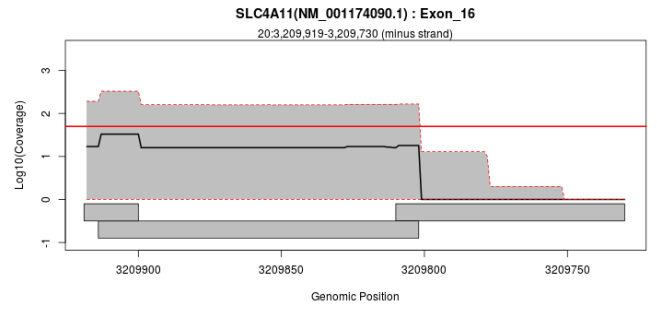
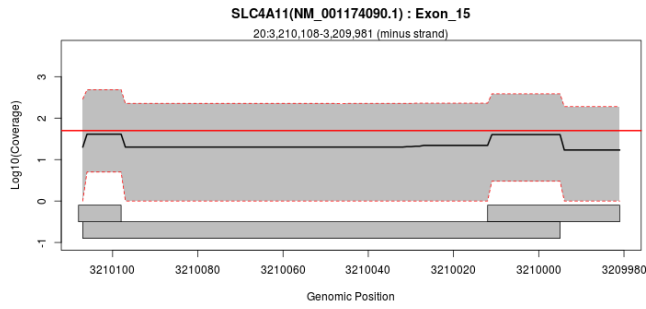


## Exon Overview: SLC4A11 : NM\_001174090.1

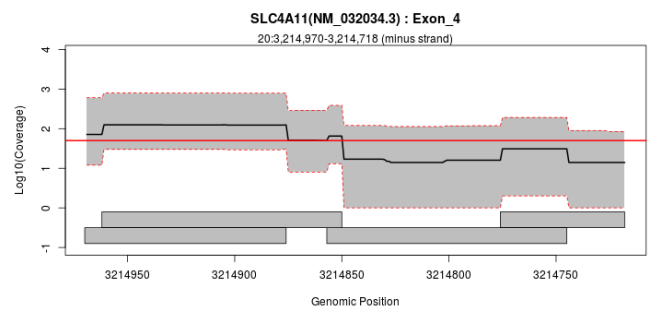
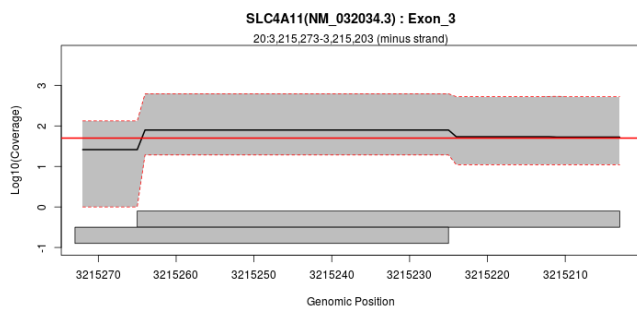
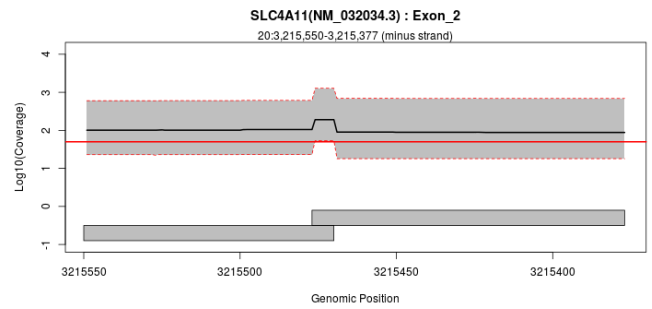
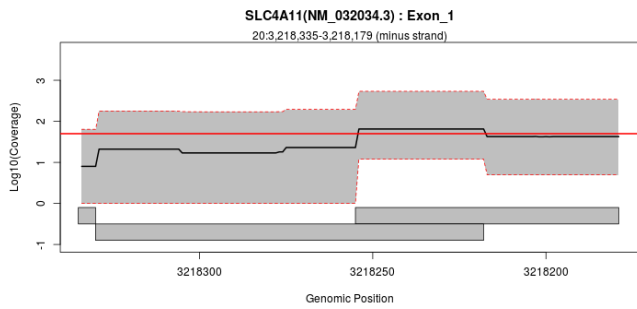


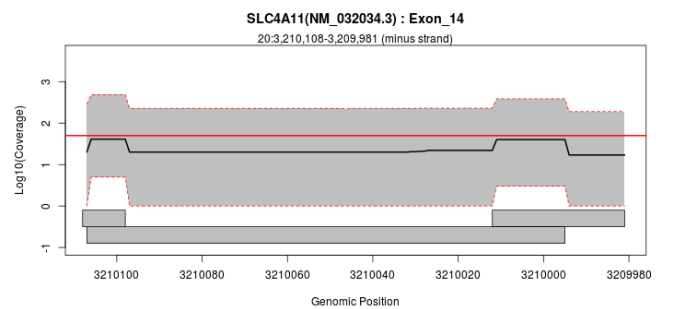
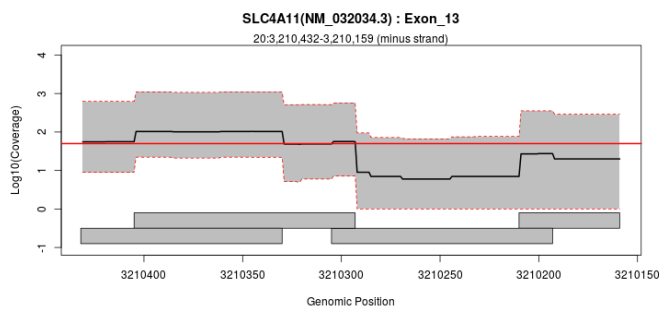
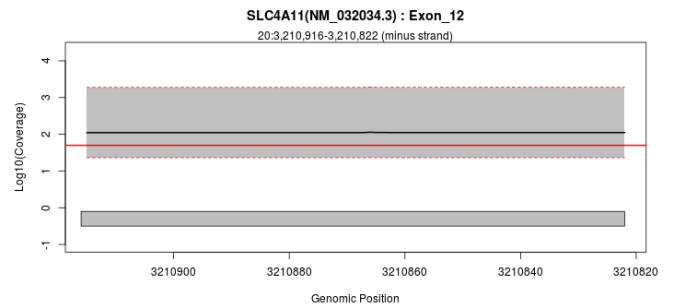
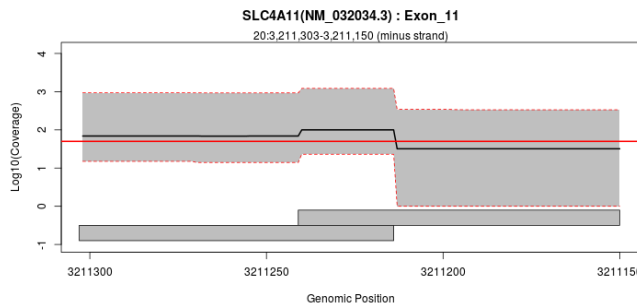
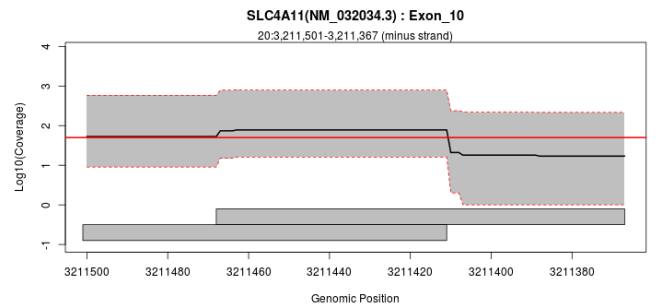
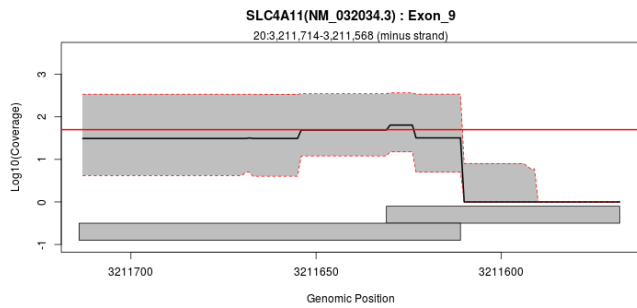
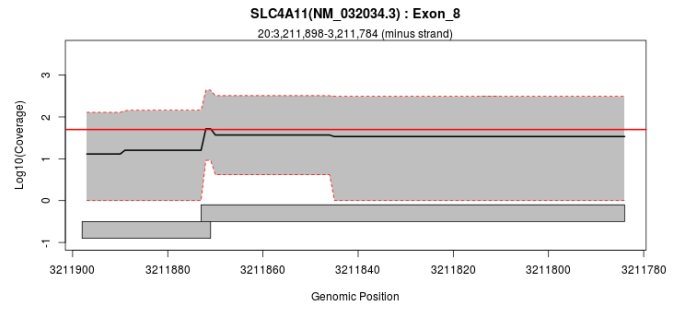
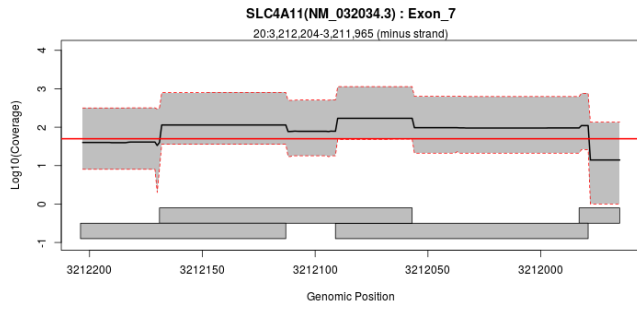
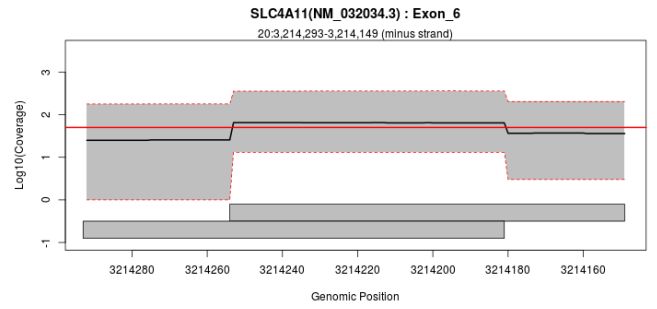
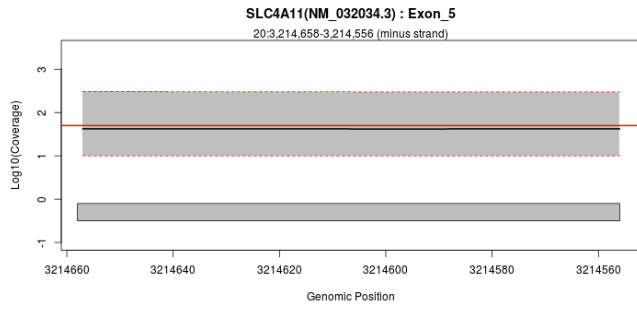


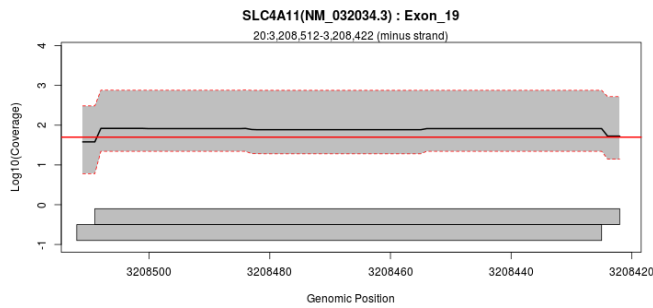
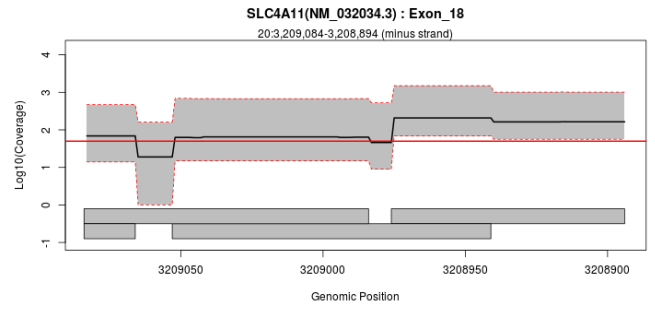
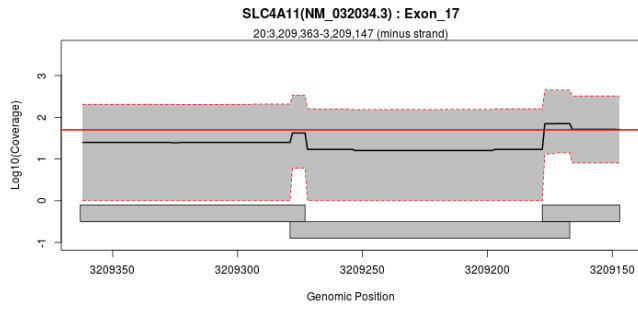
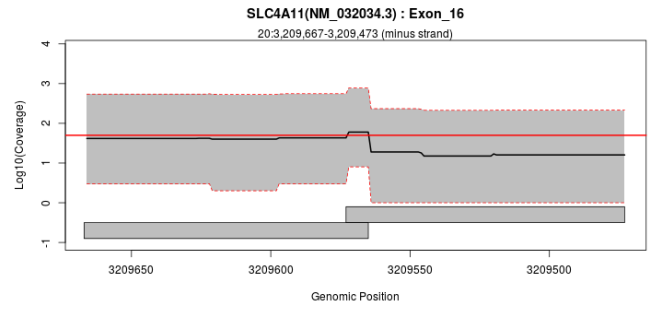
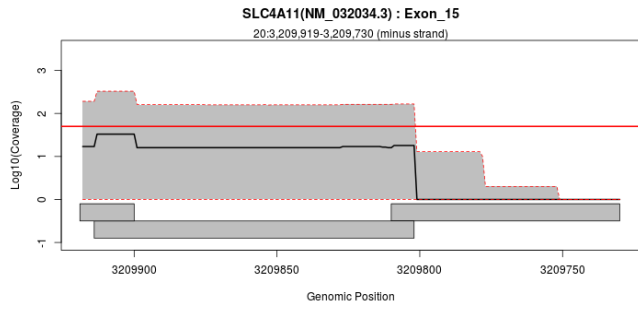




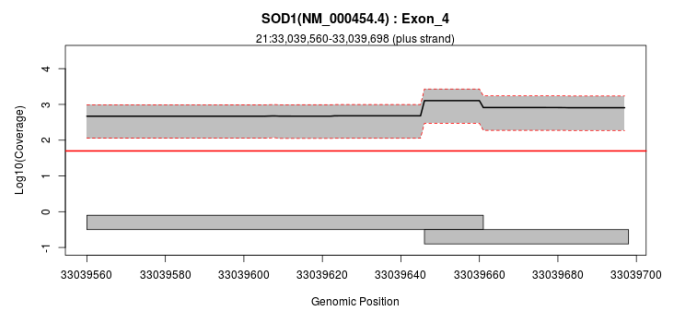
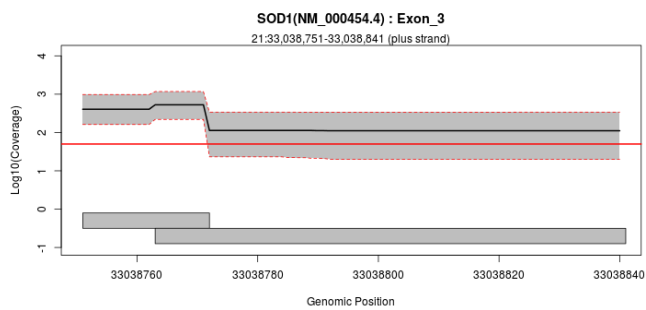
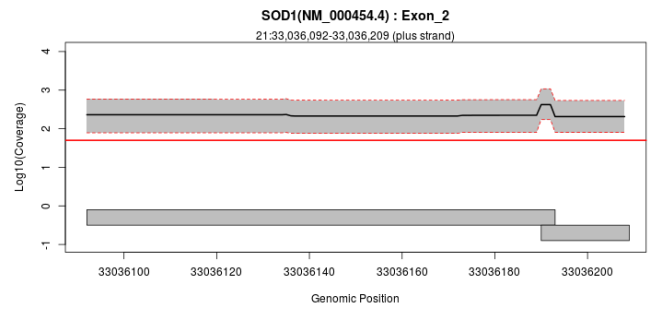
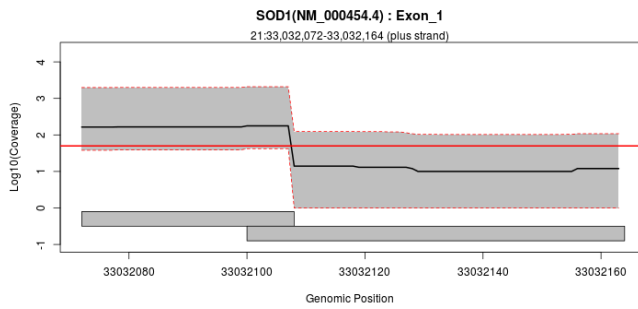
## Exon Overview: SLC4A11 : NM\_032034.3

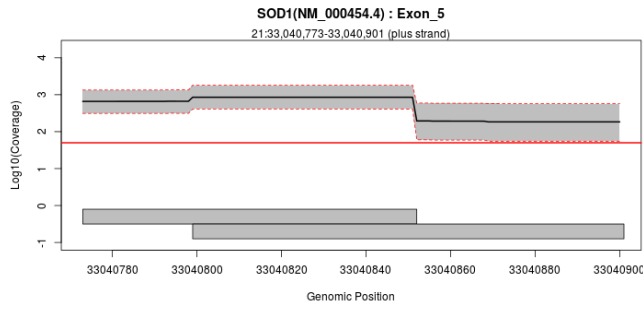




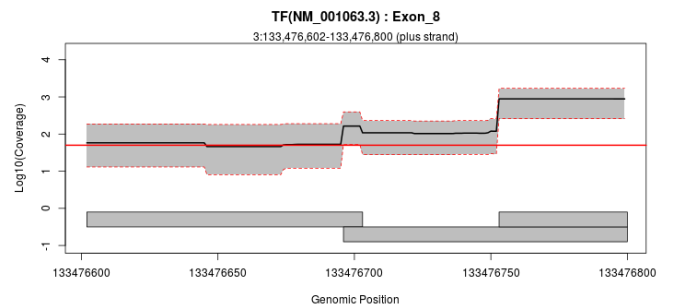
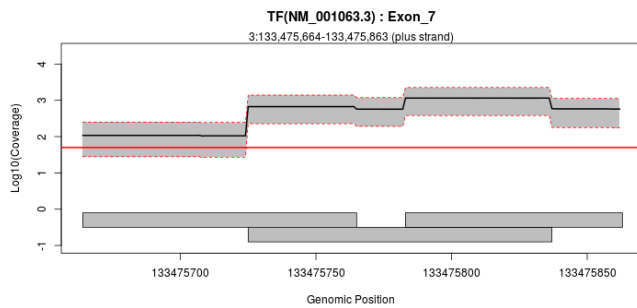
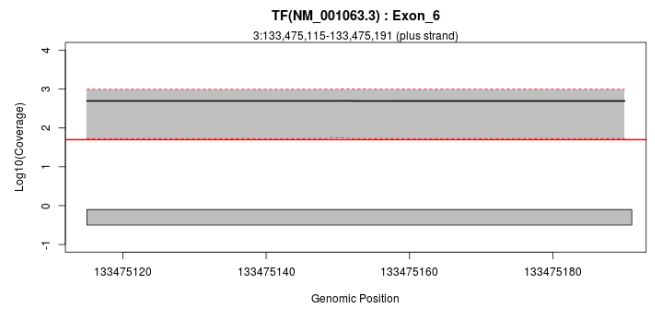
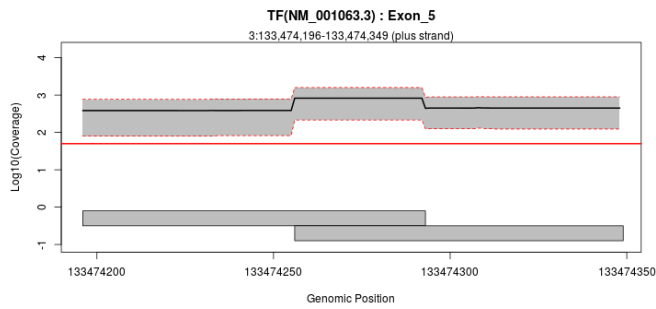
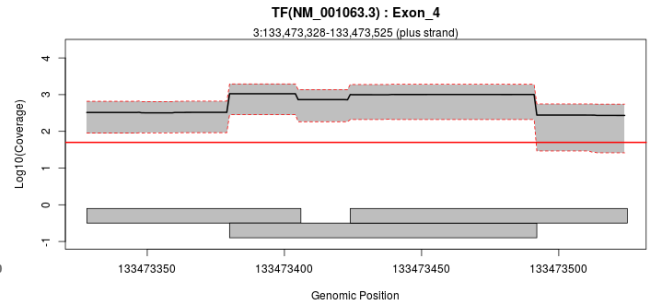
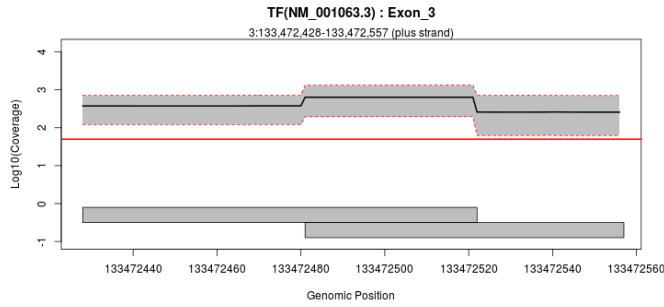
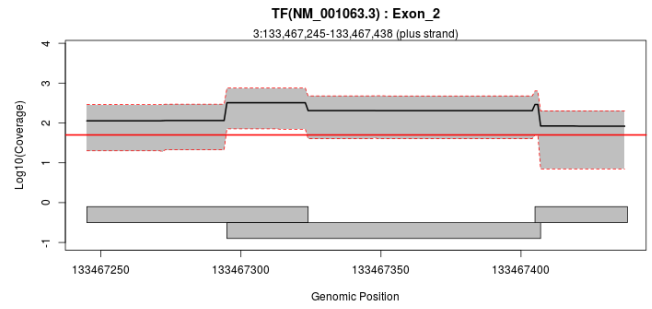
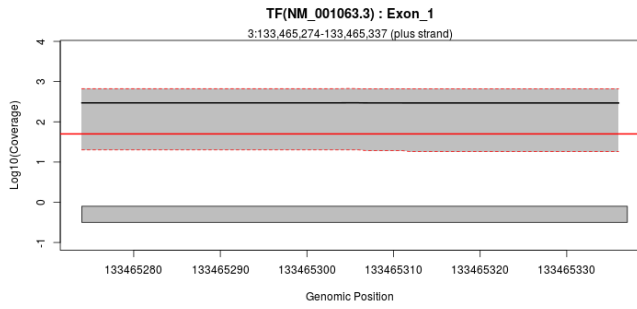


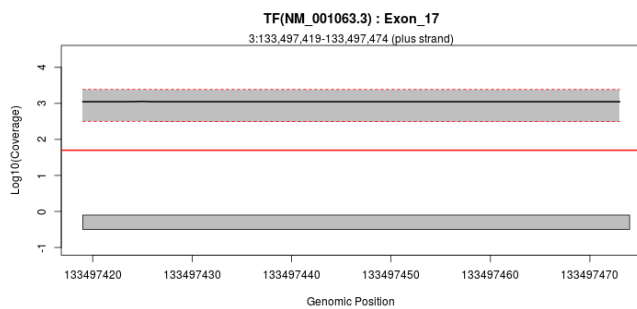
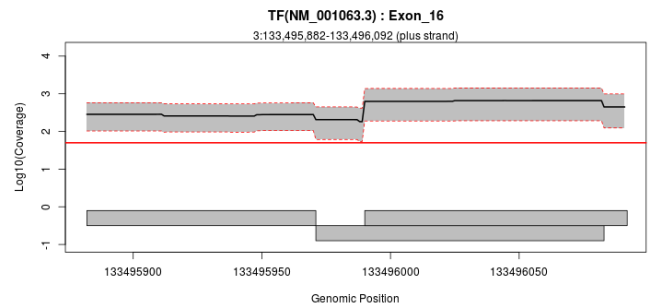
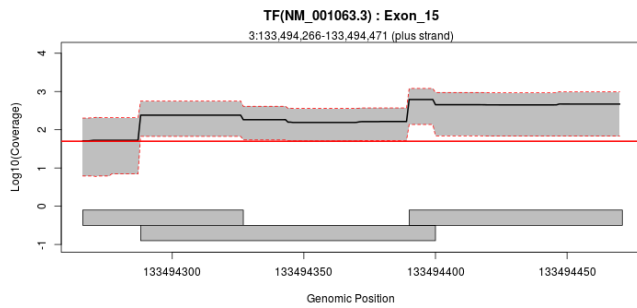
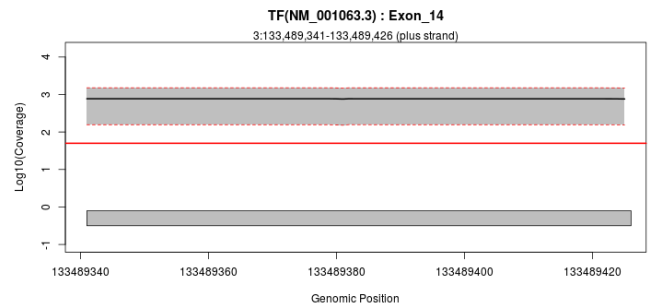
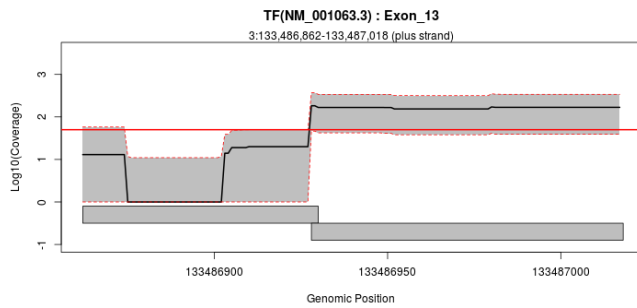
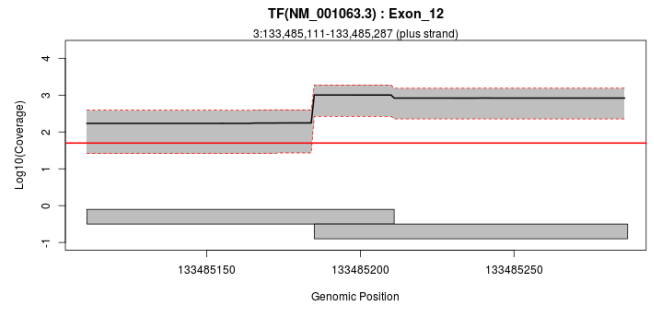
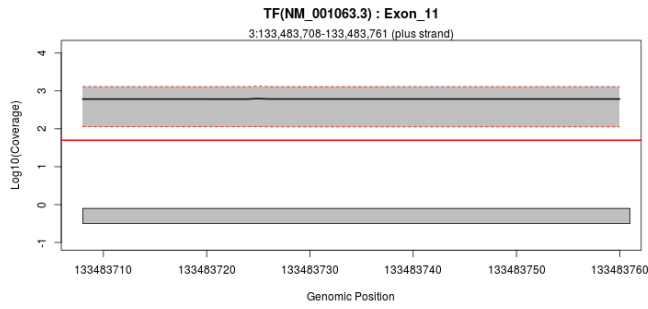
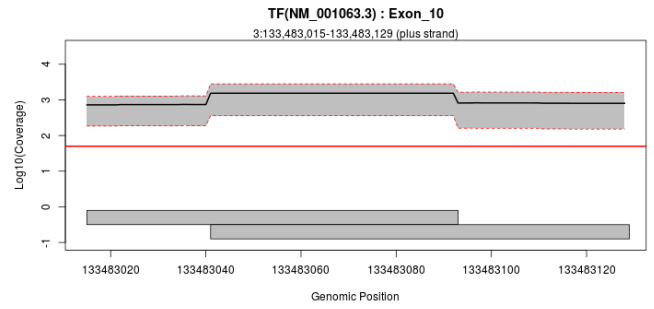
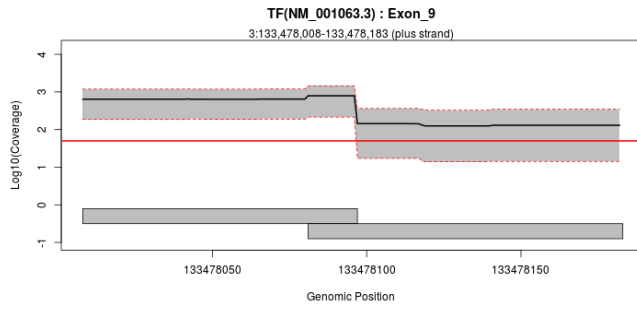
## Exon Overview: SOD1 : NM\_000454.4



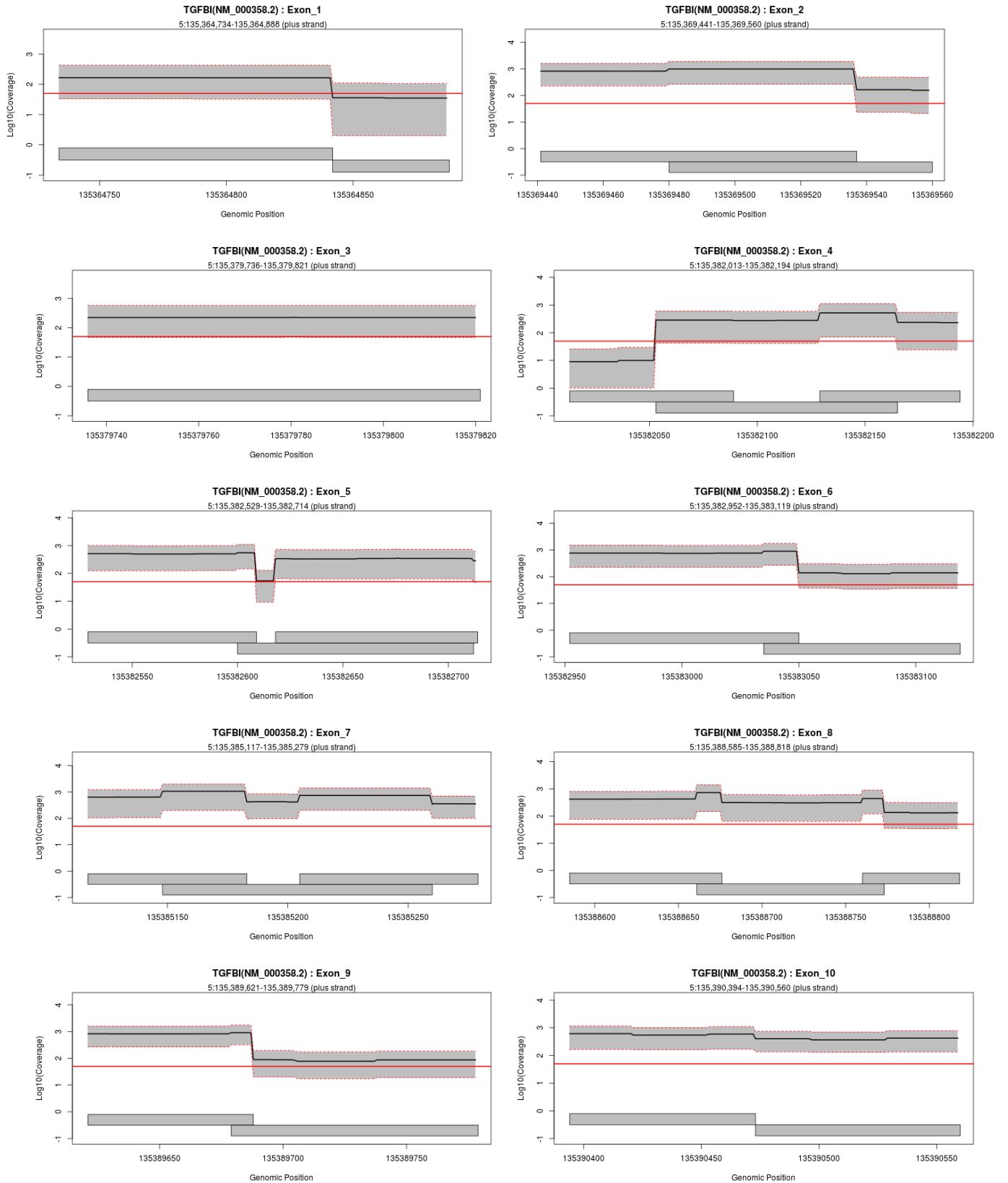


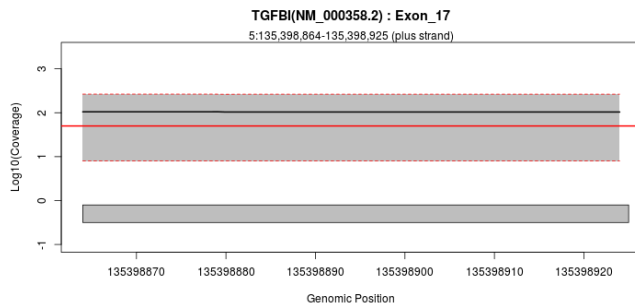
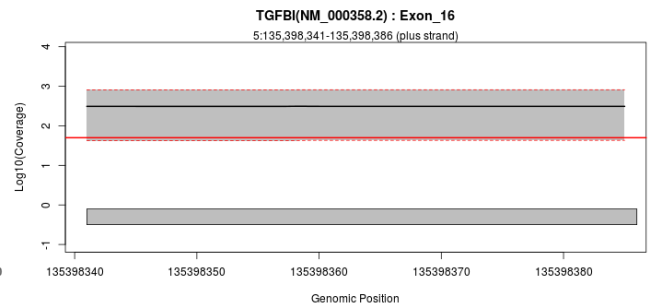
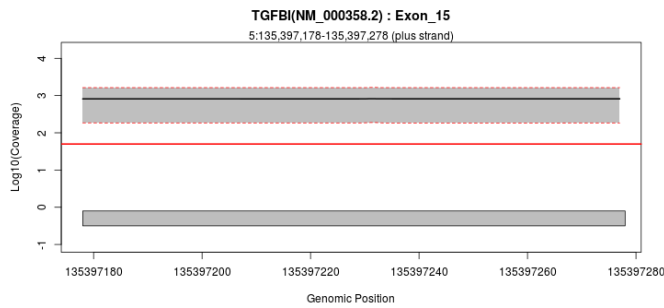
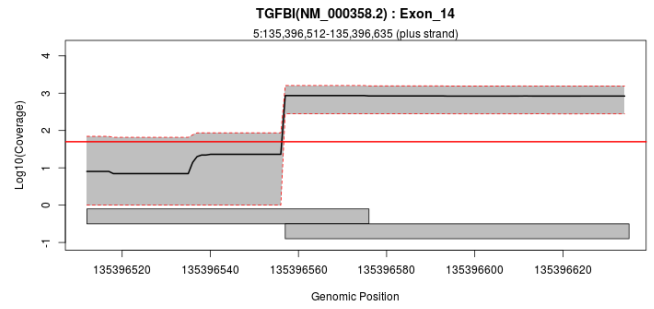
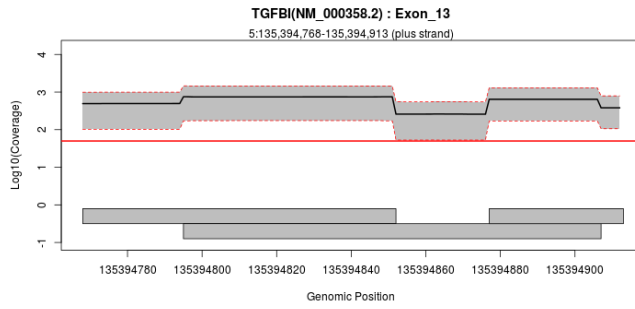
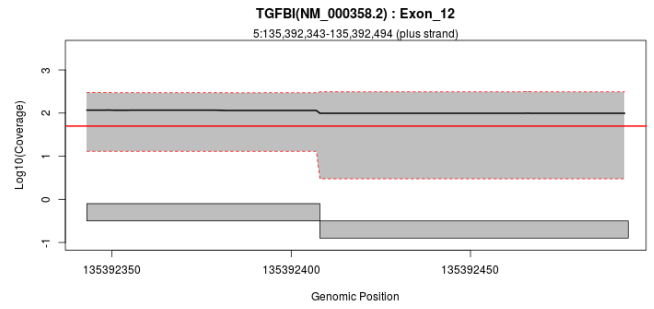
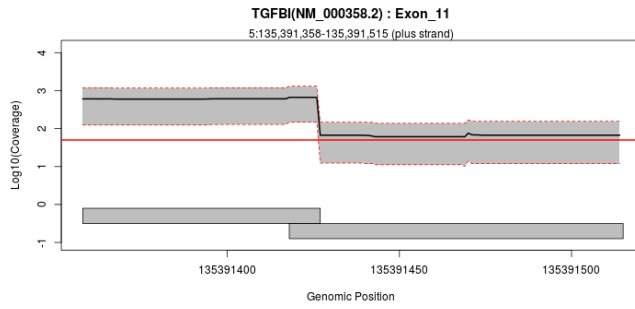
## Exon Overview: TF : NM\_001063.3



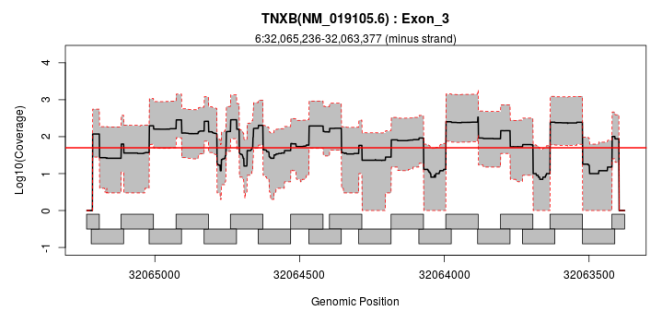
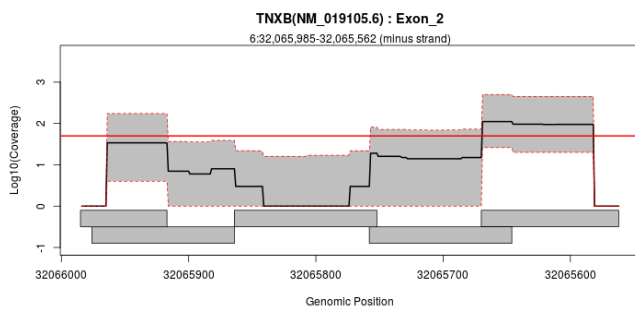


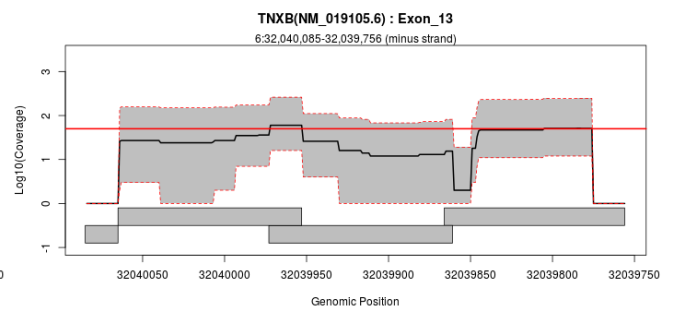
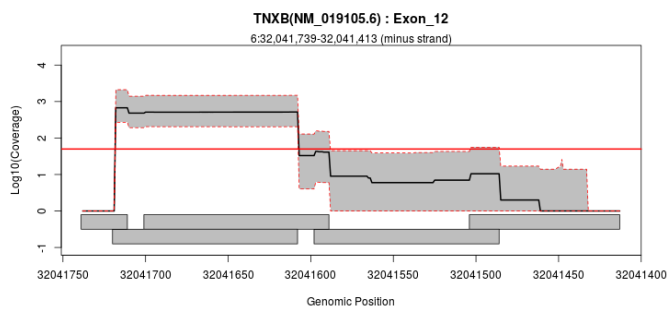
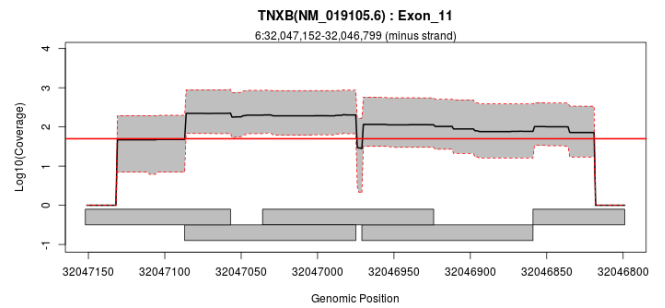
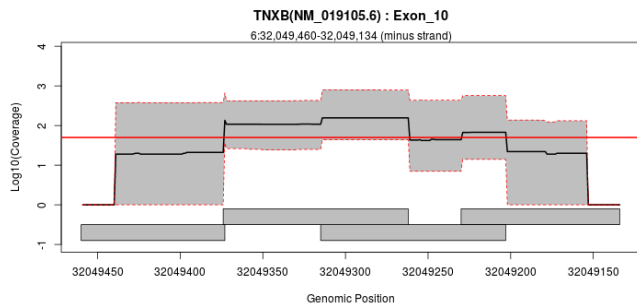
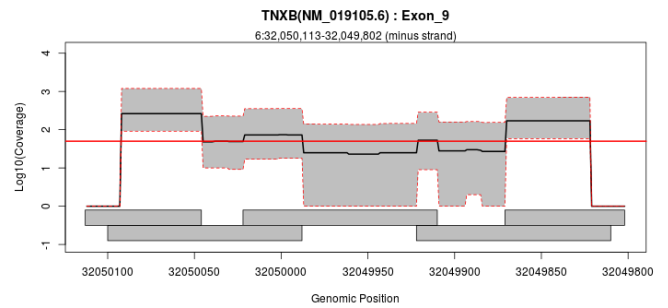
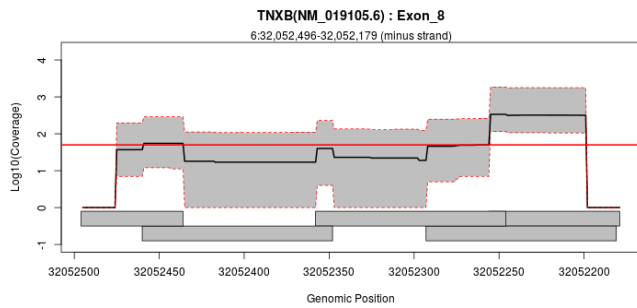
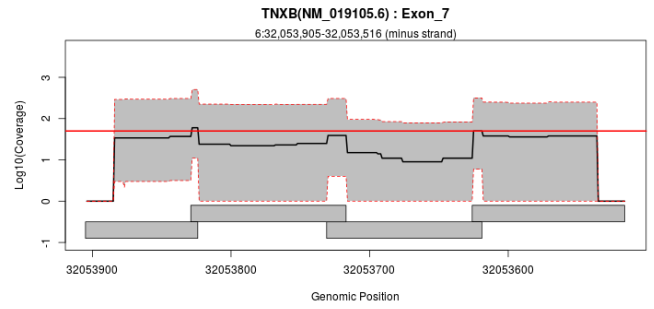
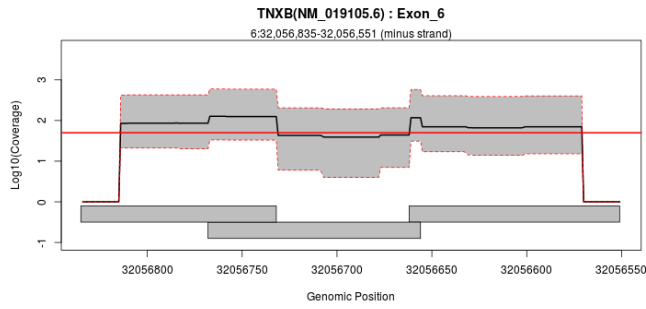
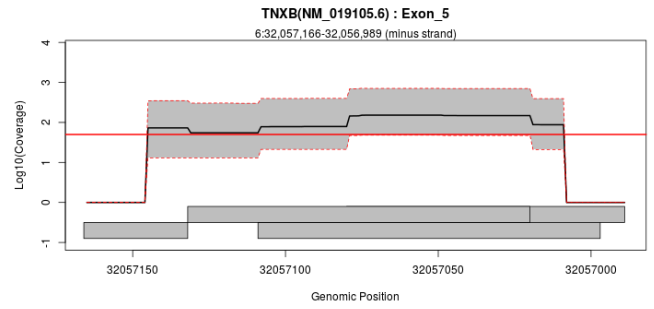
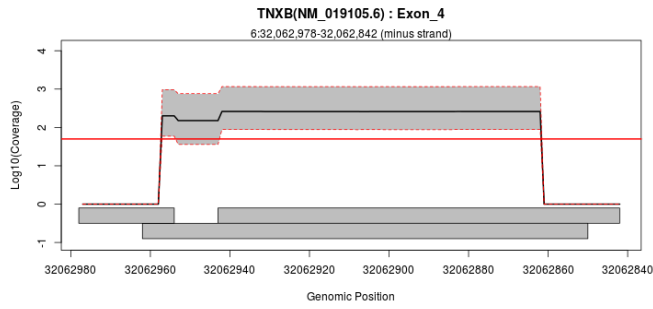
# Exon Overview: TGFBI : NM\_000358.2



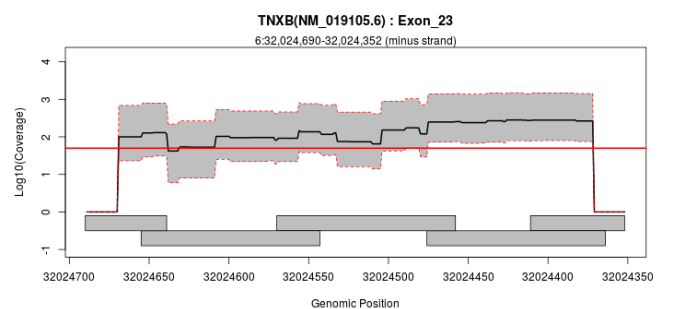
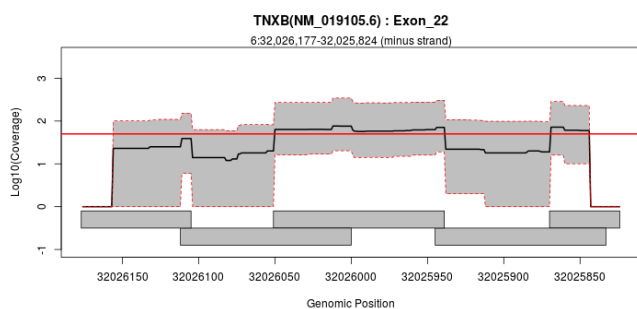
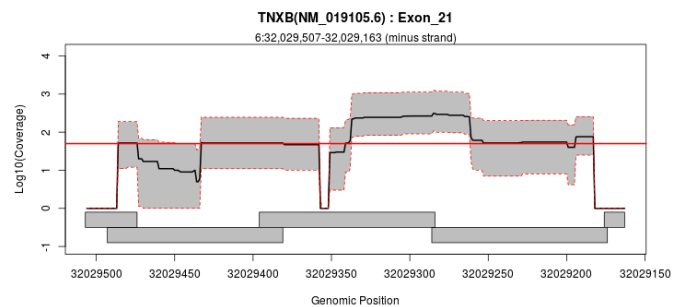
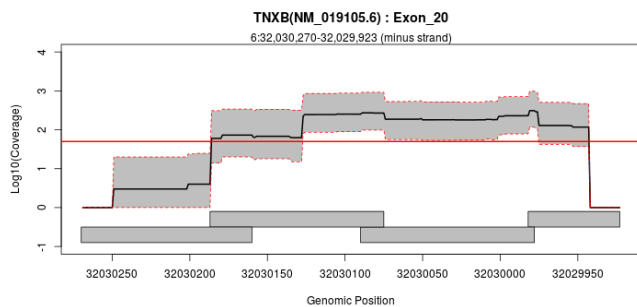
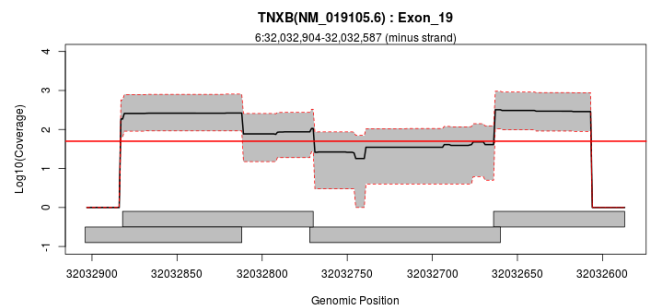
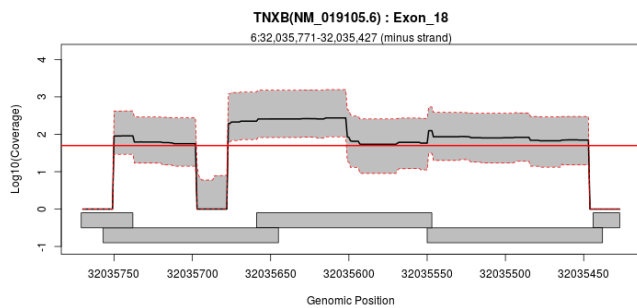
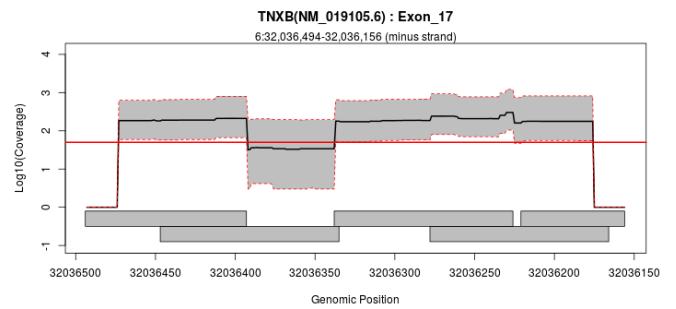
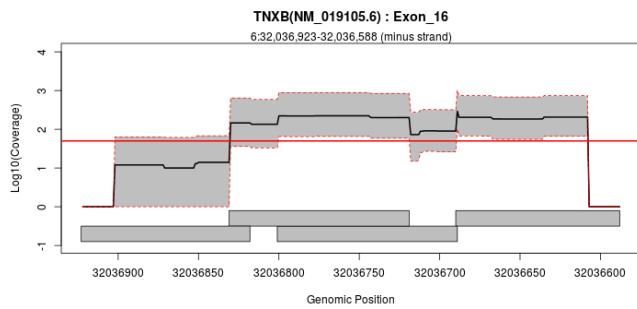
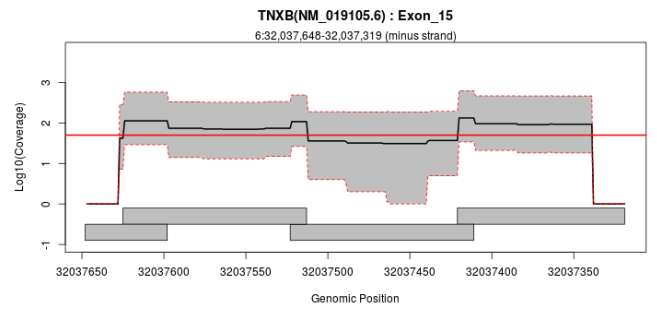
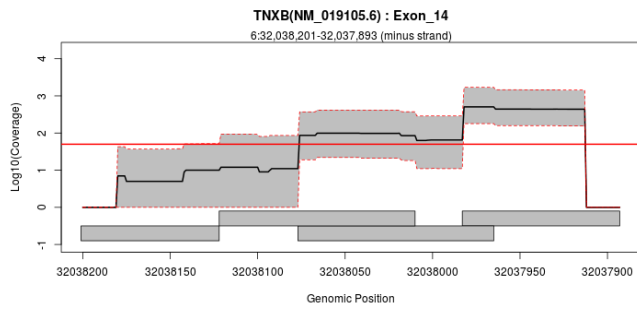


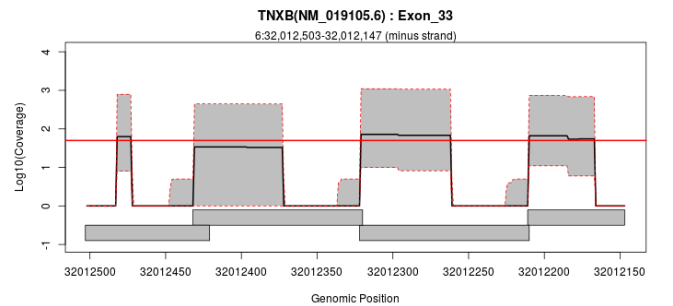
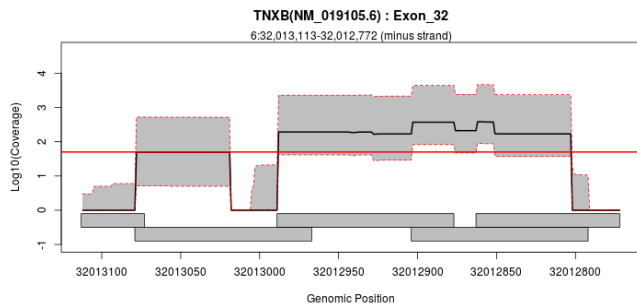
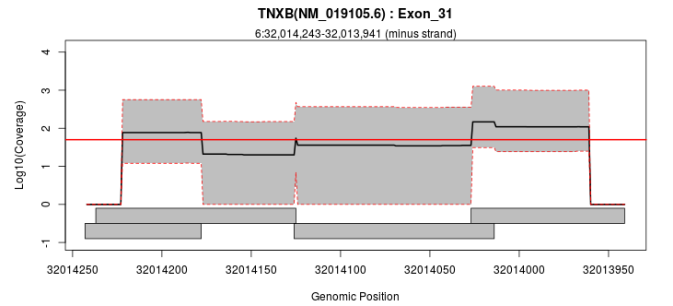
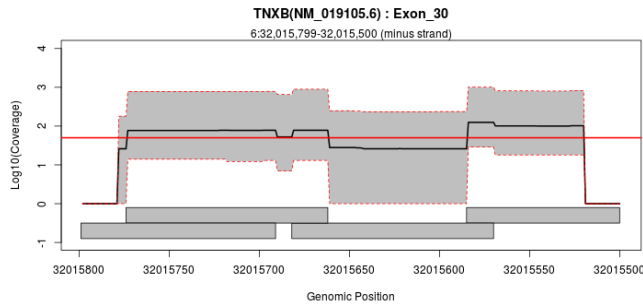
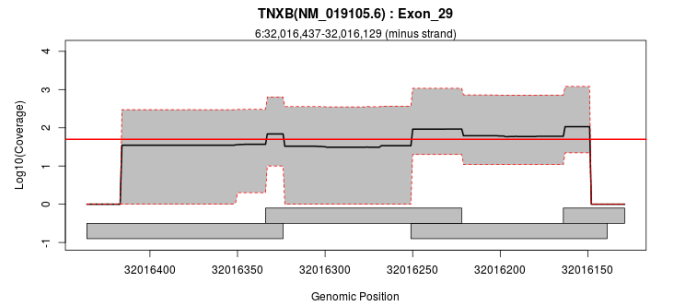
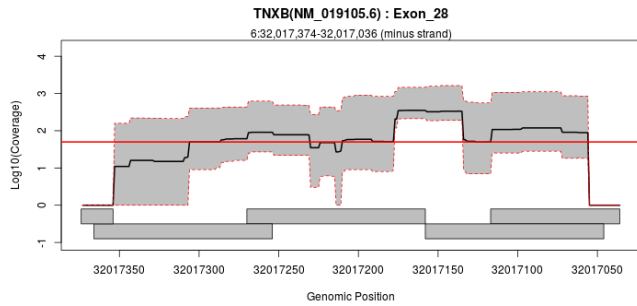
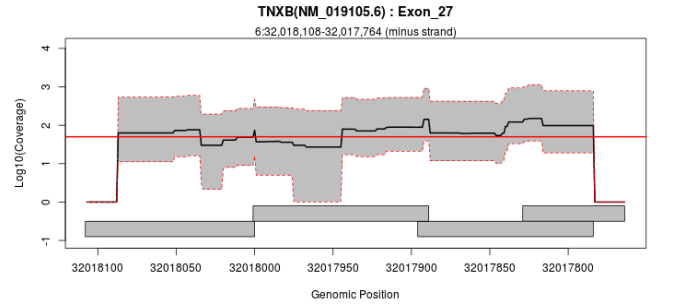
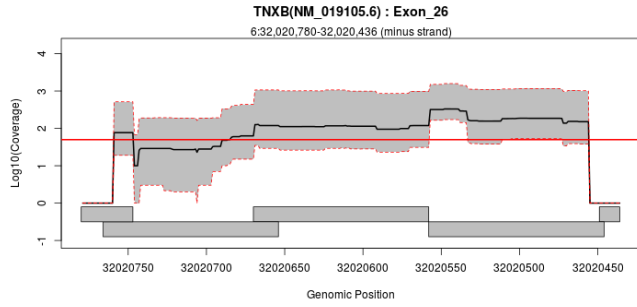
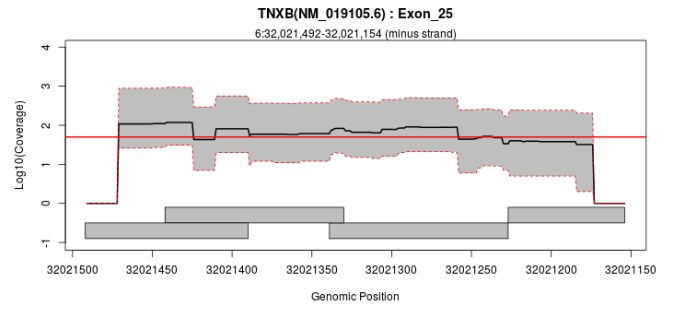
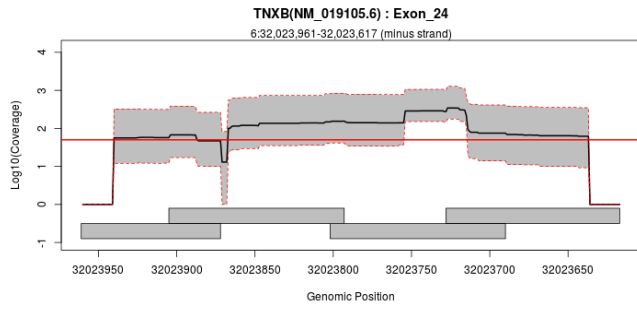
## Exon Overview: TNXB : NM\_019105.6

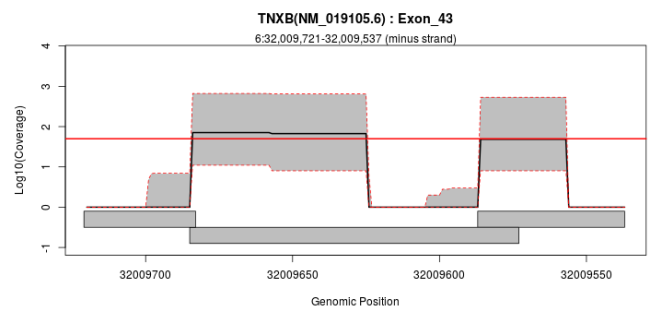
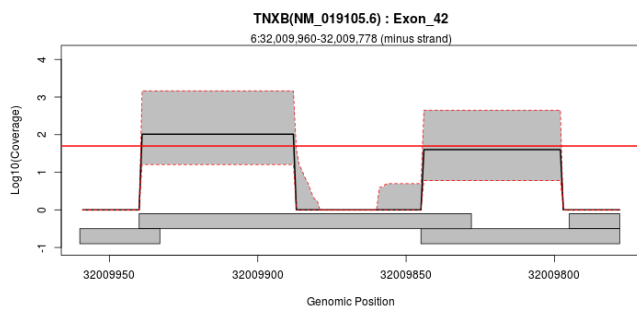
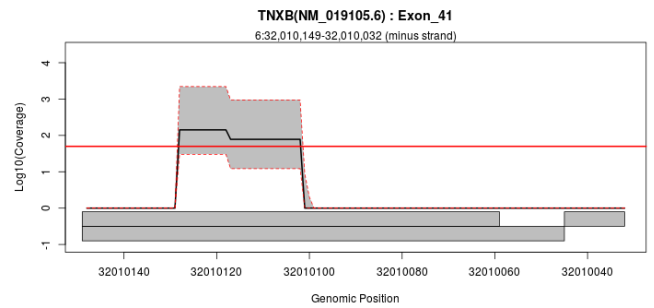
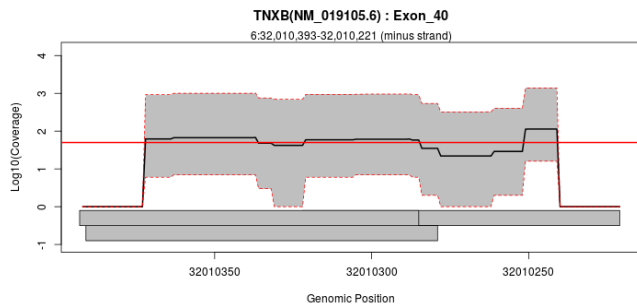
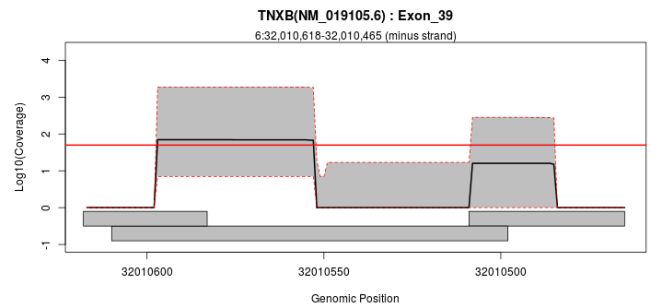
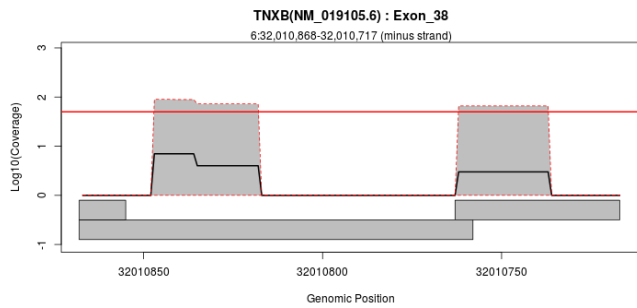
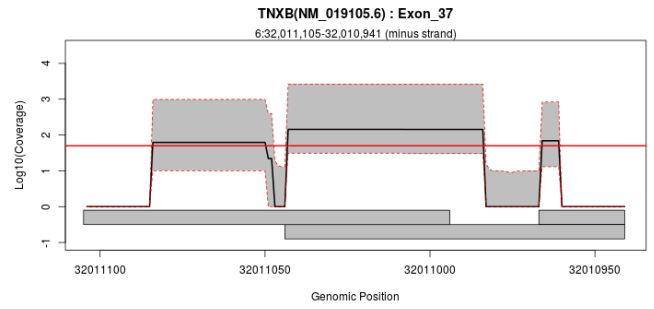
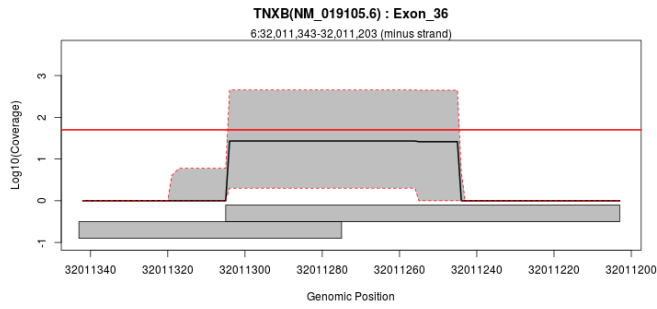
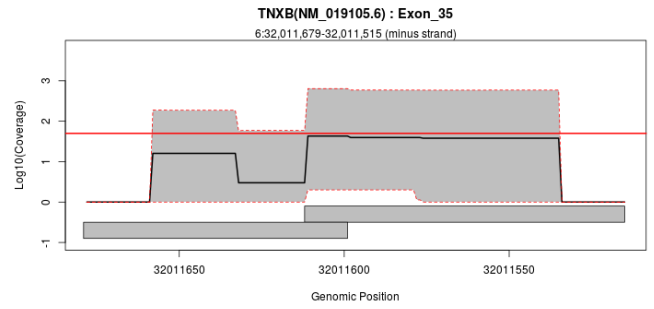
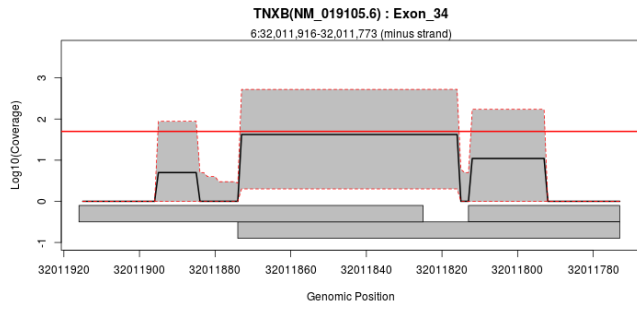


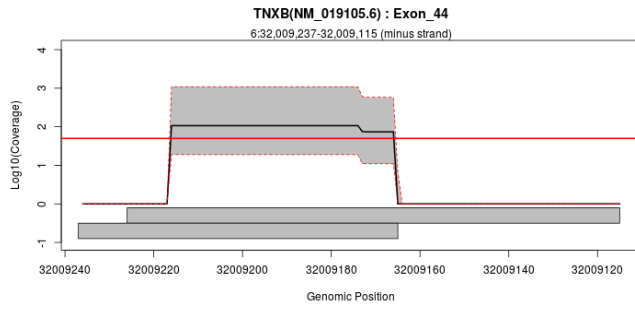




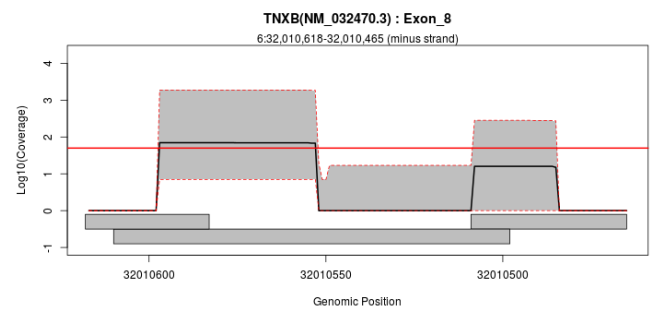
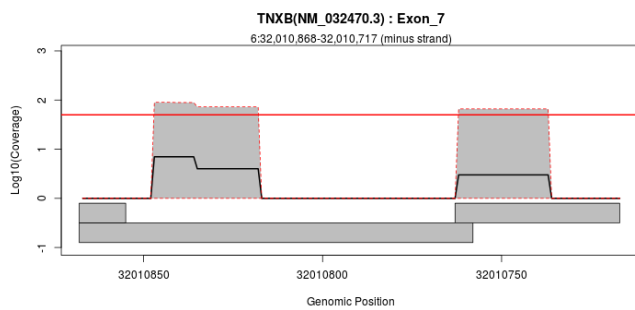
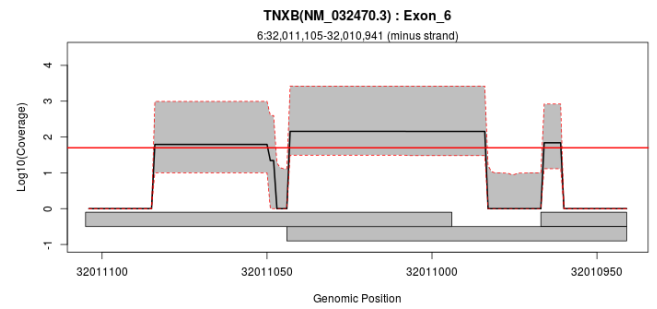
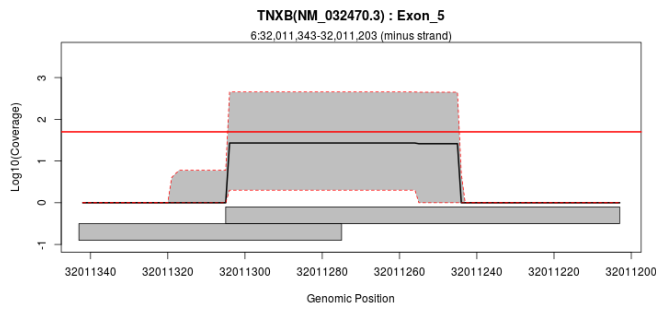
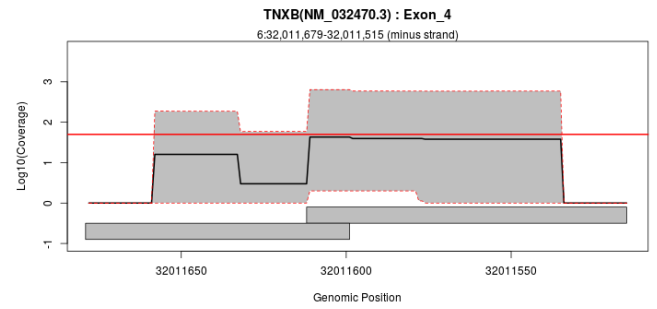
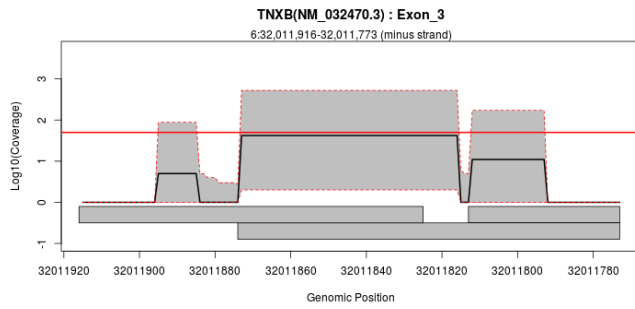
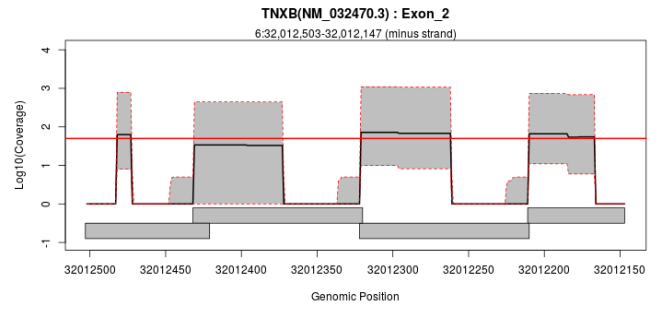
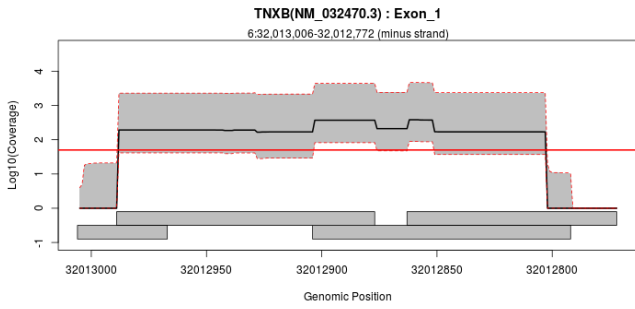


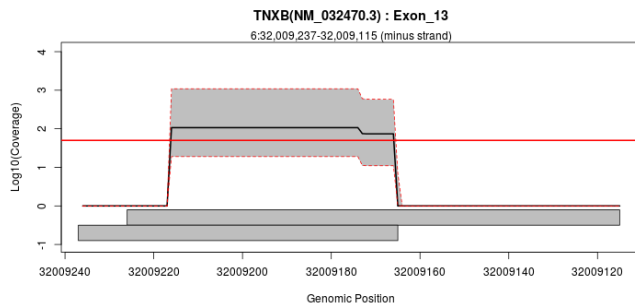
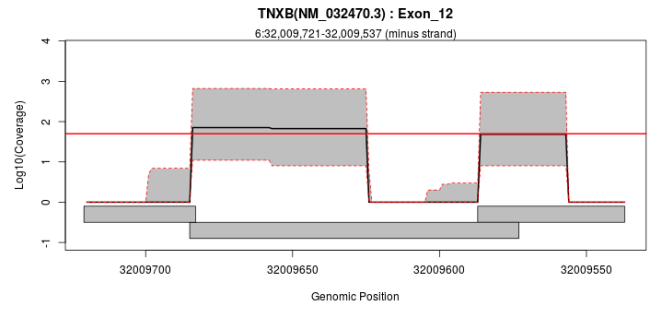
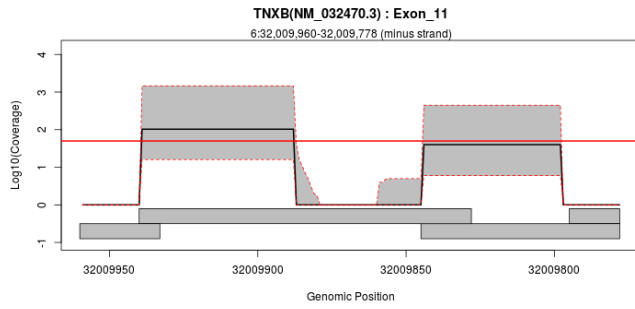
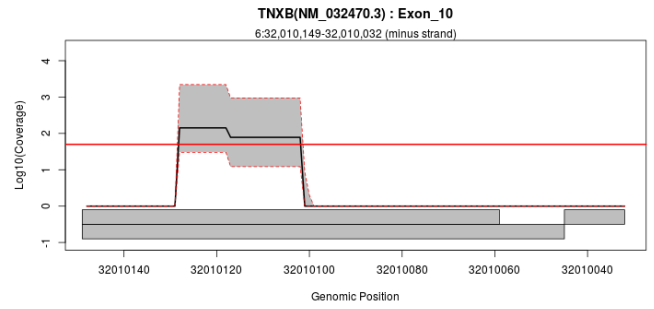
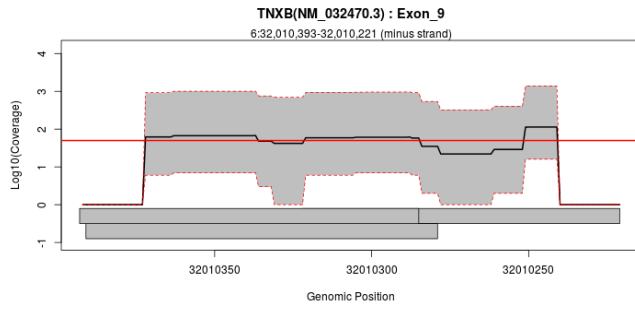




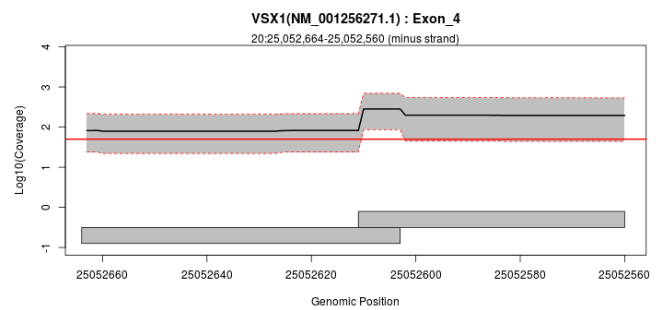
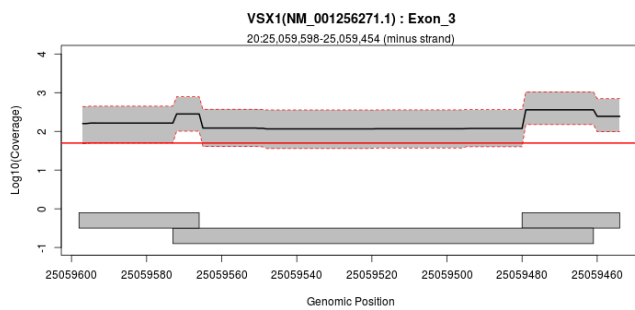
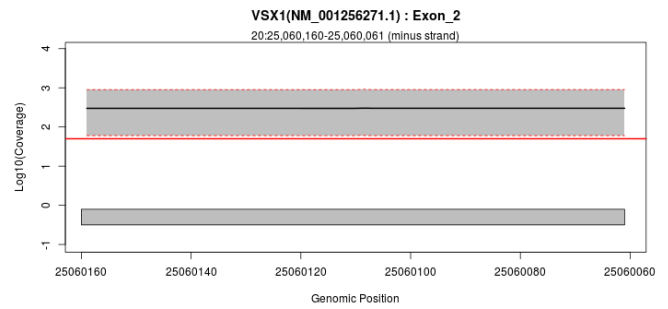
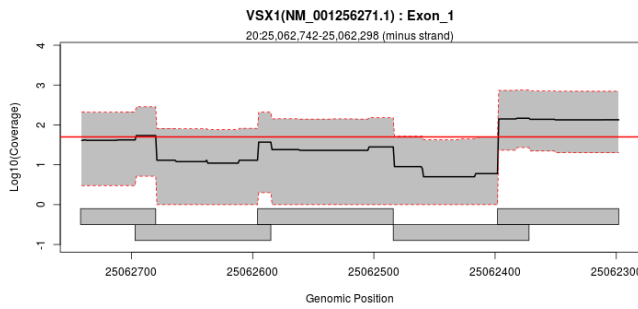


## Exon Overview: TNXB : NM\_032470.3

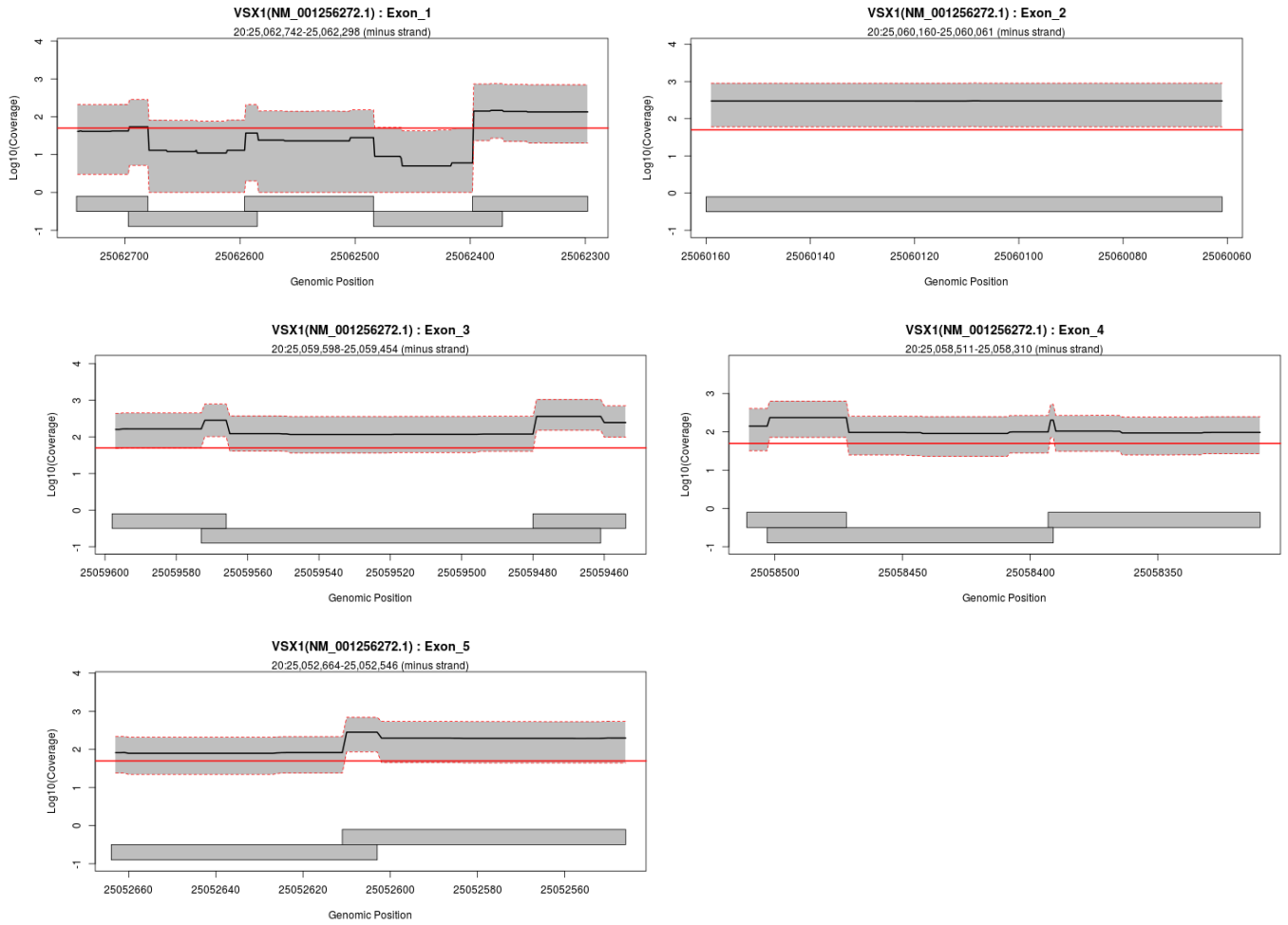




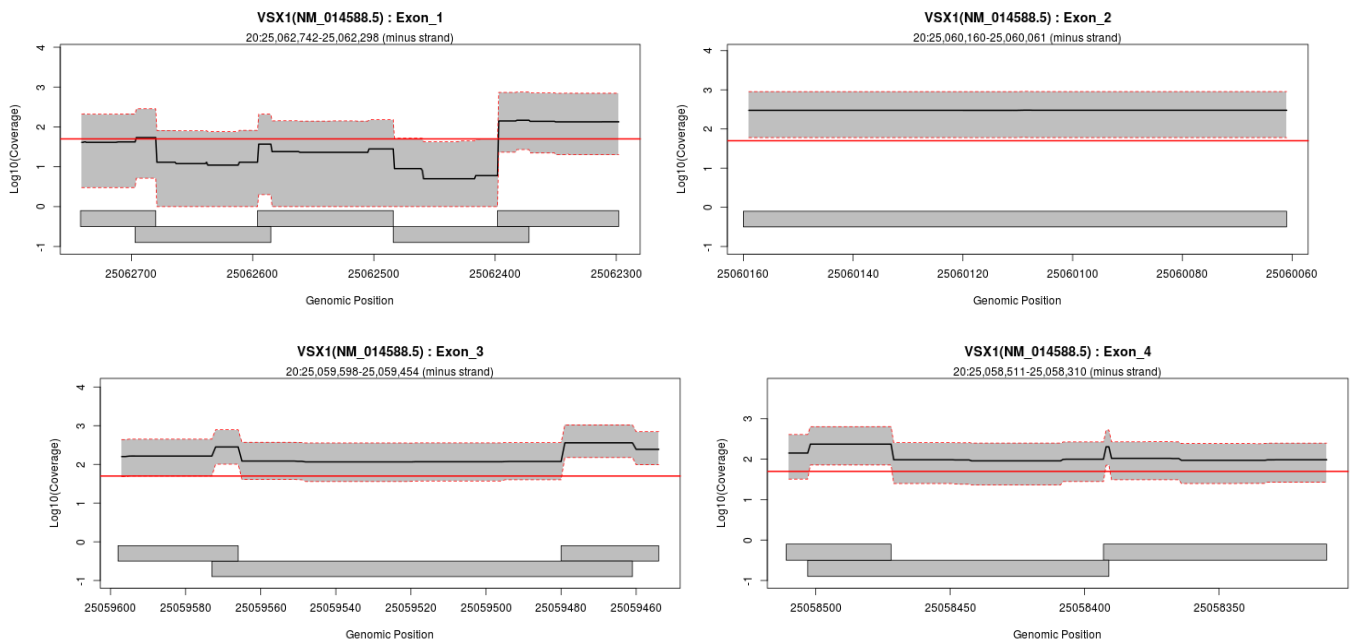
## Exon Overview: VSX1 : NM\_001256271.1

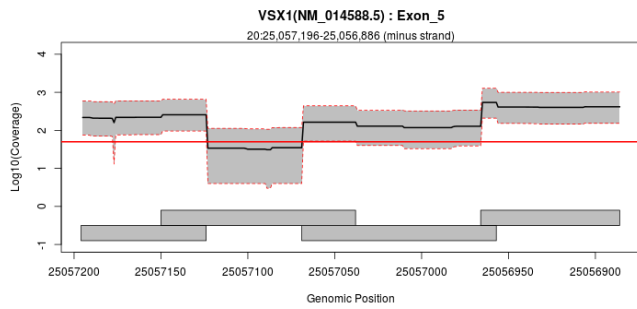


## Exon Overview: VSX1 : NM\_001256272.1

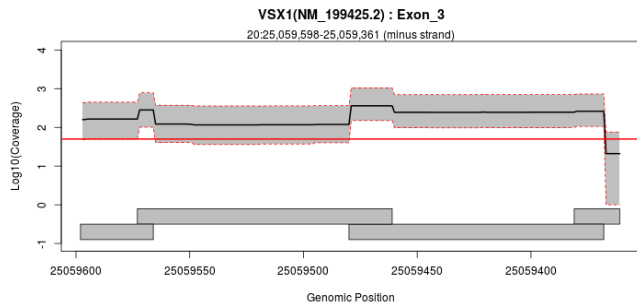
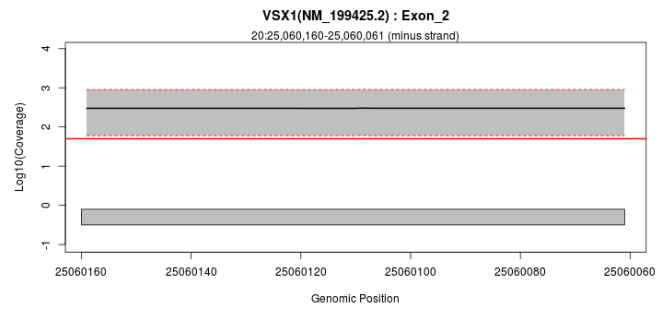
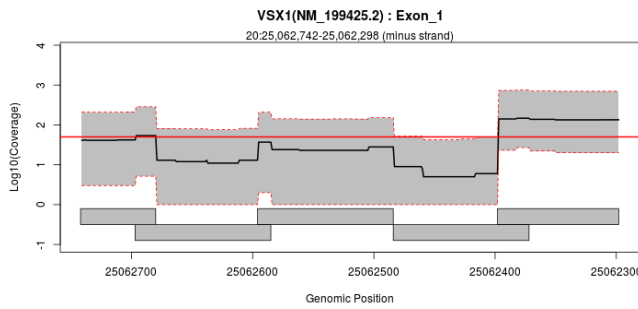


## Exon Overview: VSX1 : NM\_014588.5

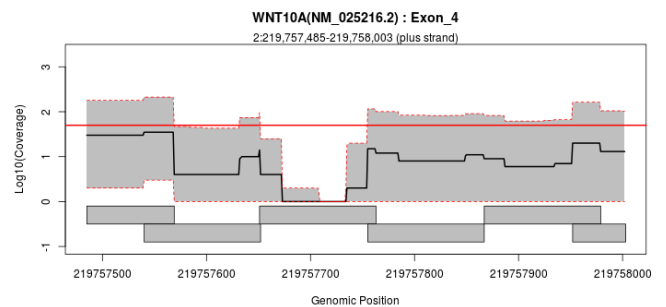
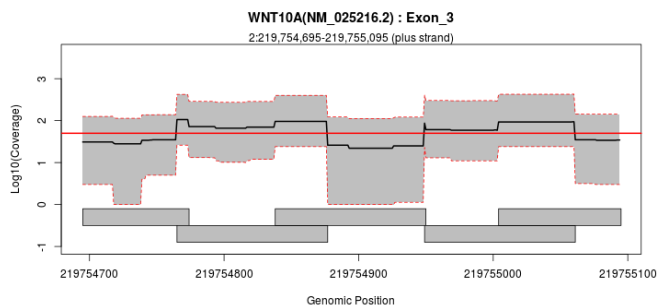
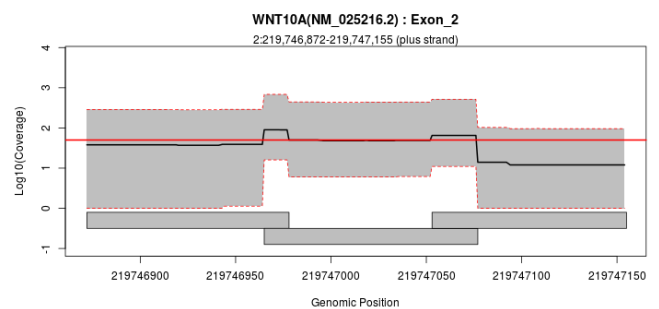
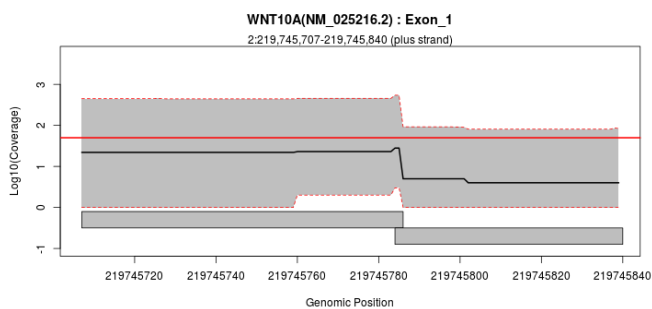




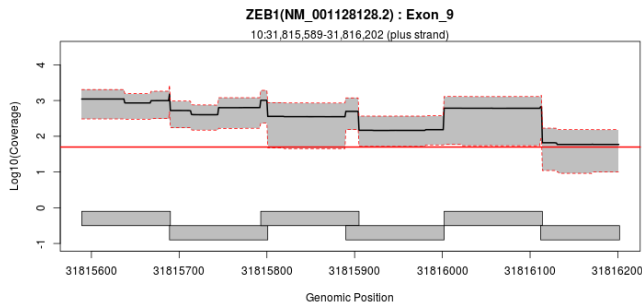
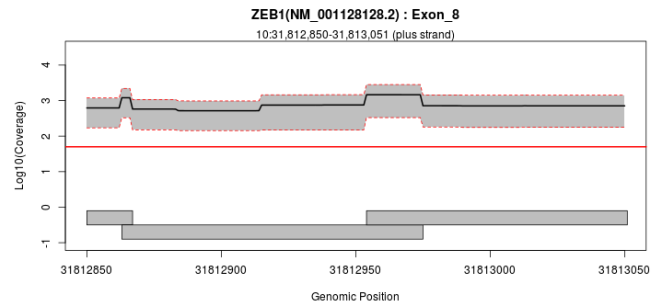
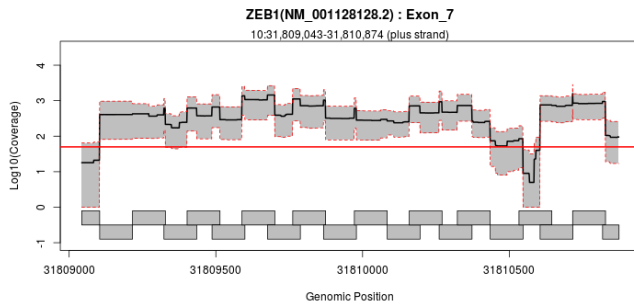
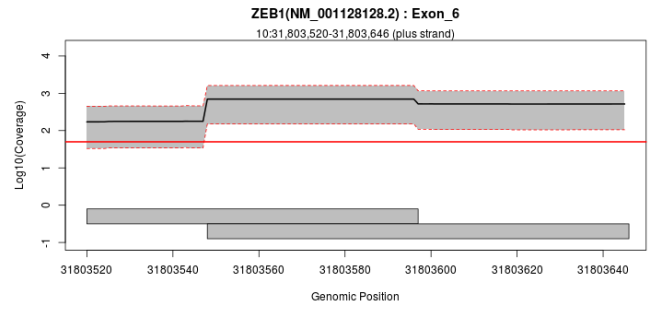
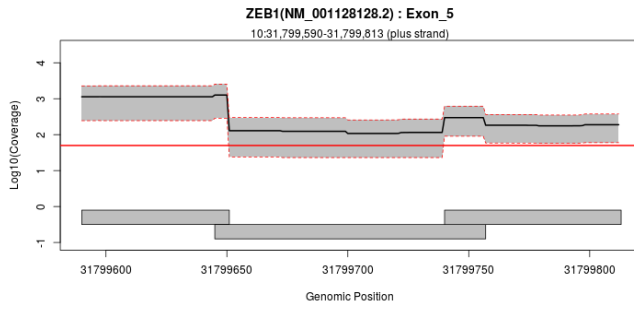
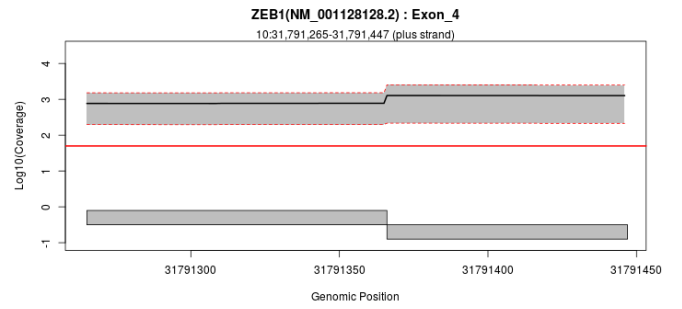
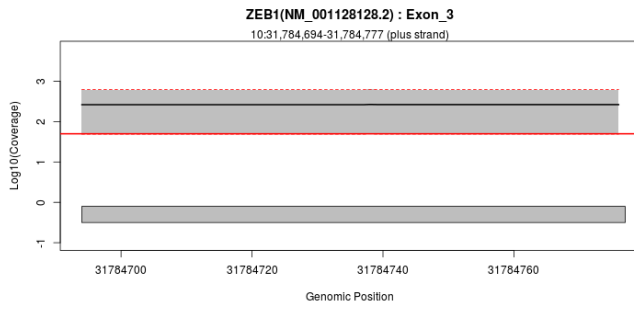
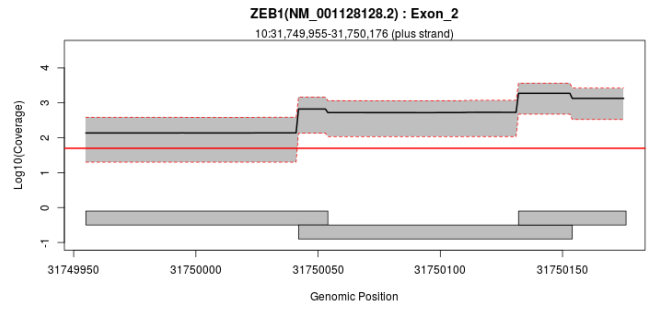
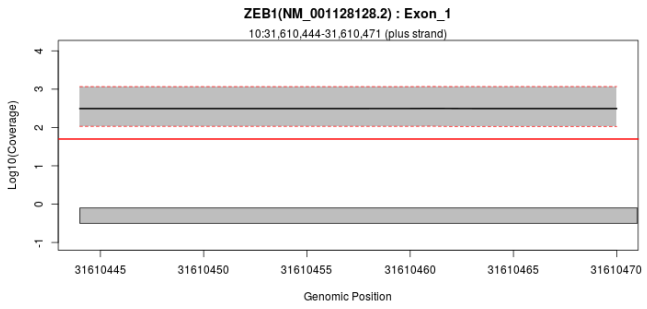
## Exon Overview: VSX1 : NM\_199425.2



## Exon Overview: WNT10A : NM\_025216.2

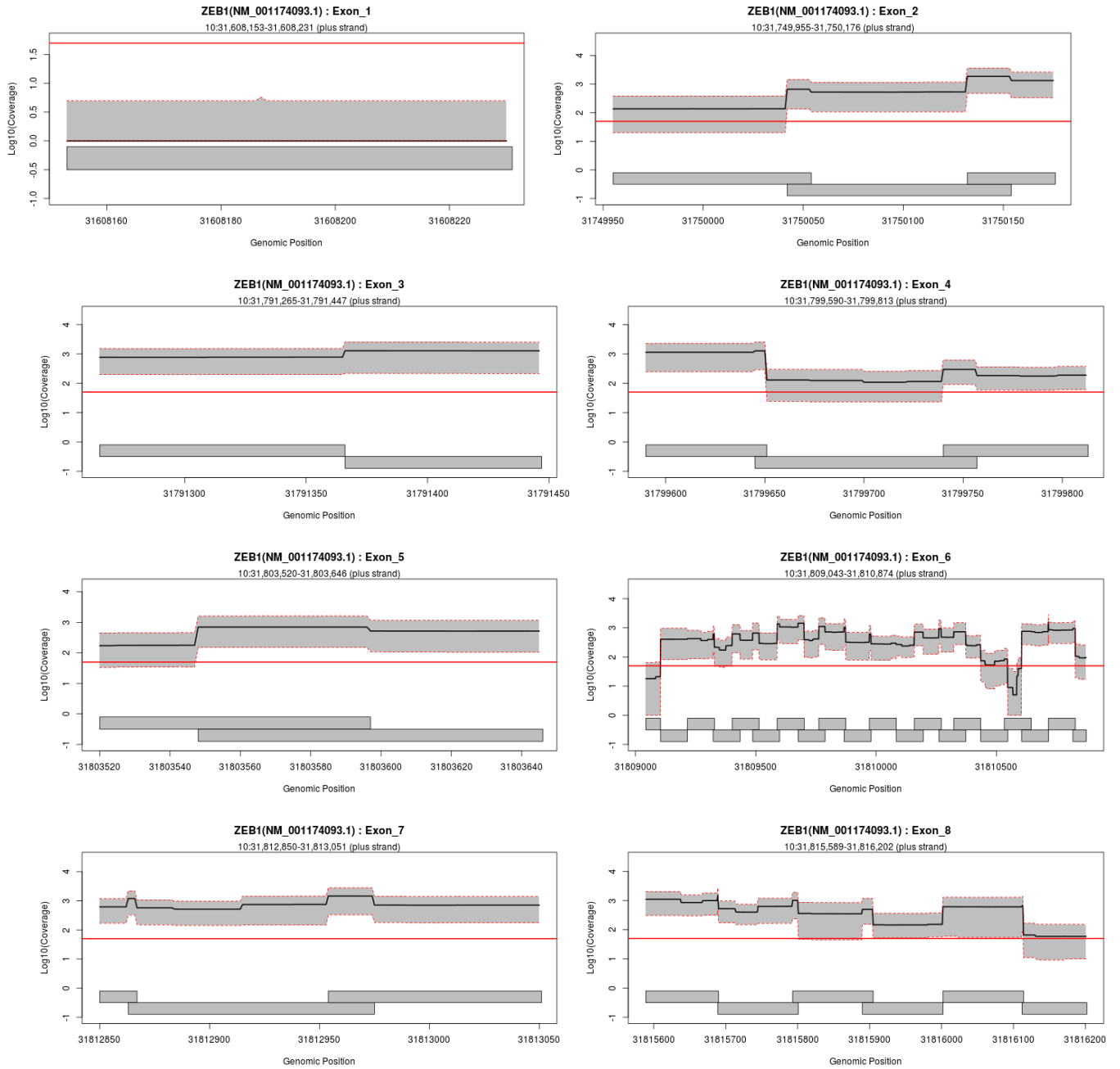


# Exon Overview: ZEB1 : NM\_001128128.2

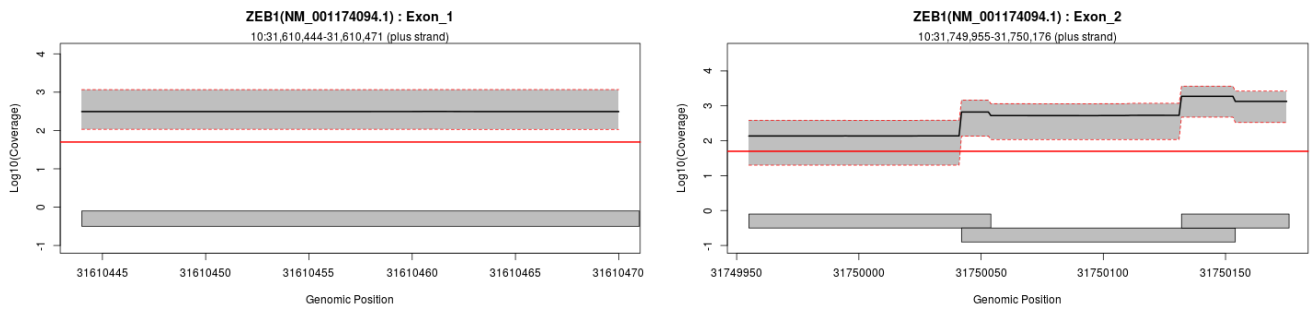


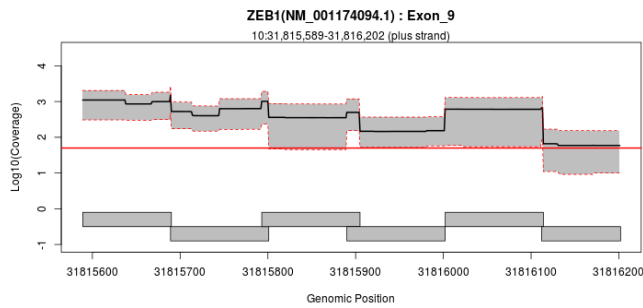
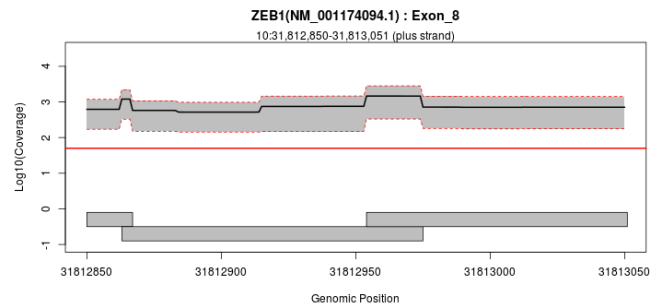
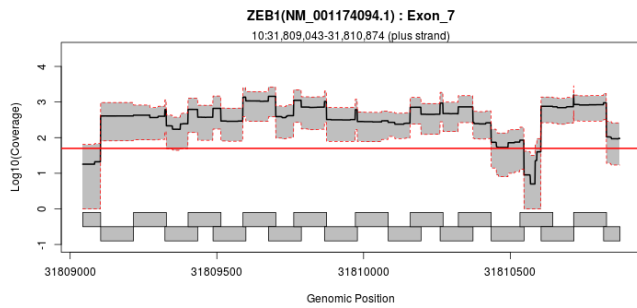
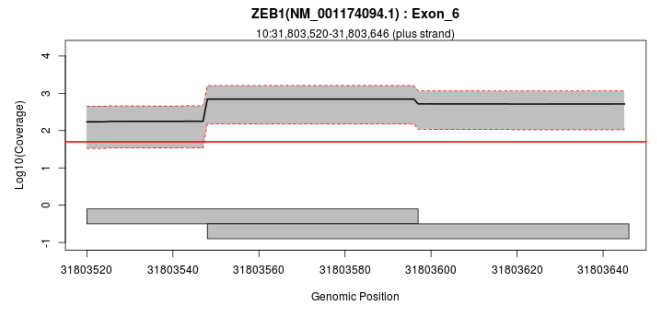
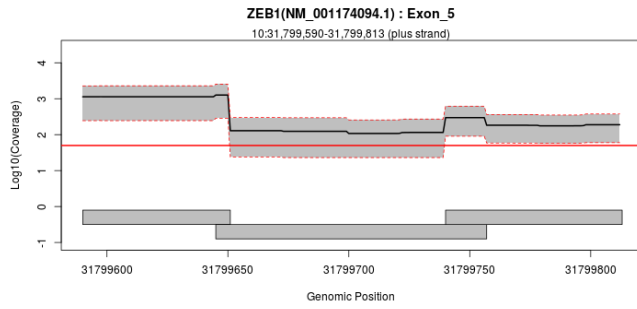
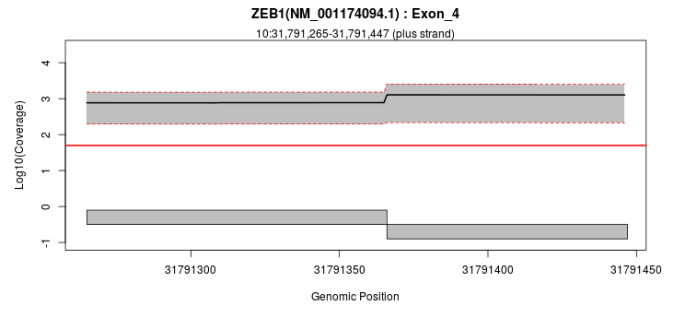
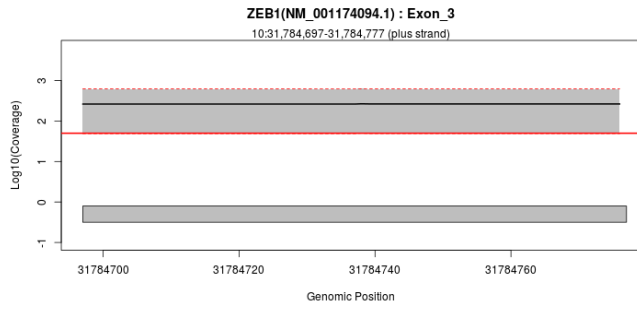


## Exon Overview: ZEB1 : NM\_001174093.1

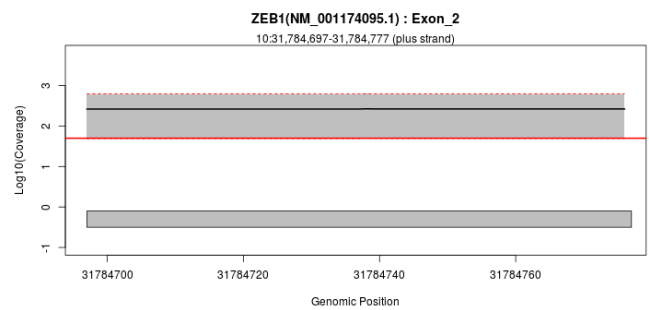
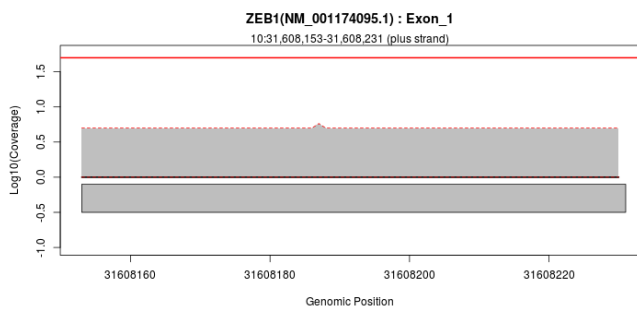


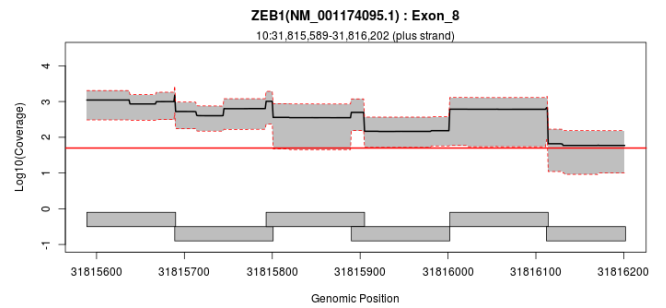
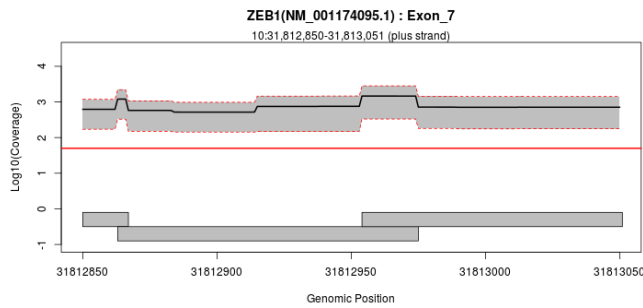
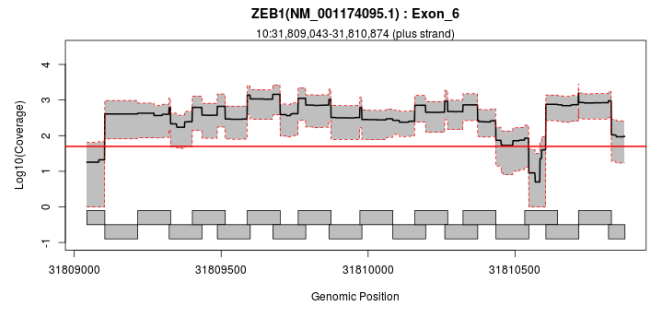
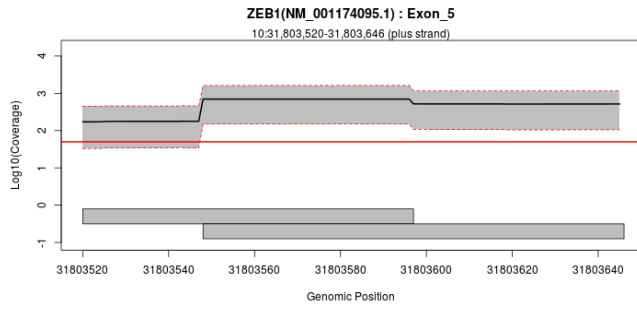
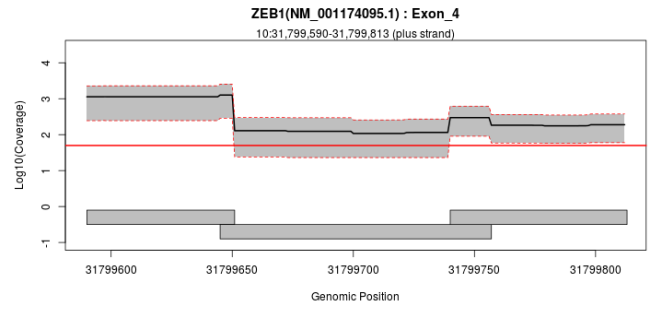
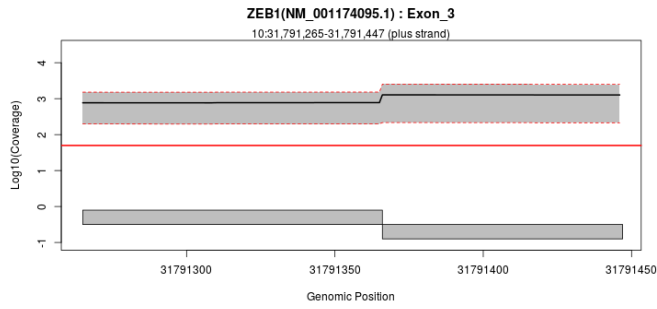
## Exon Overview: ZEB1 : NM\_001174094.1



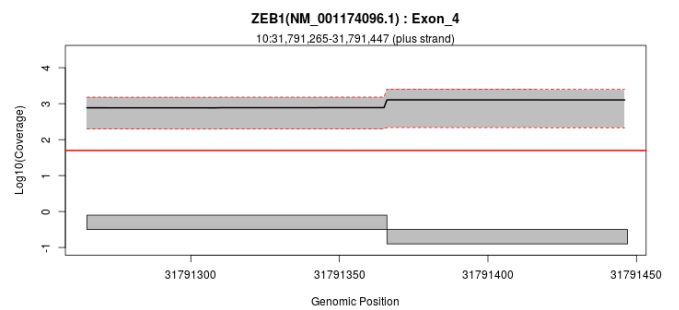
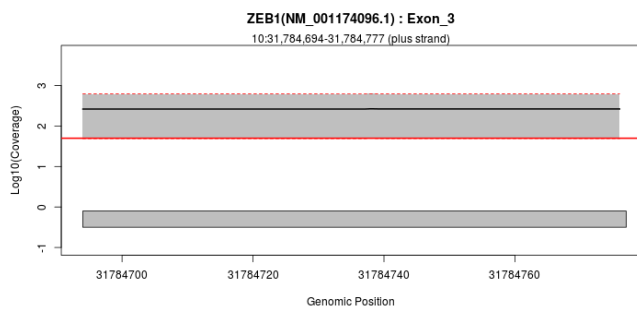
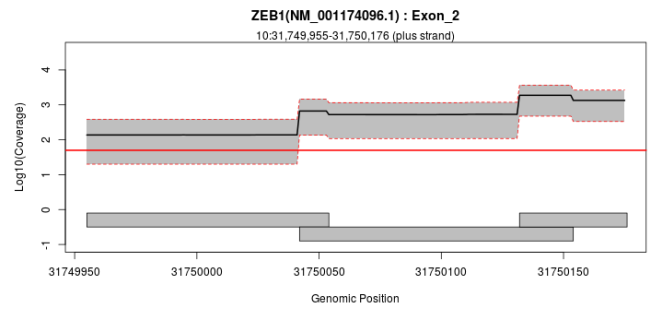
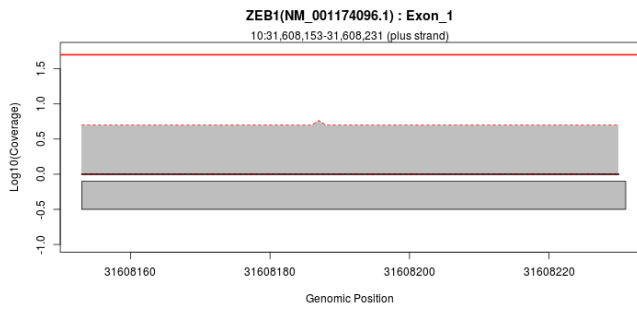


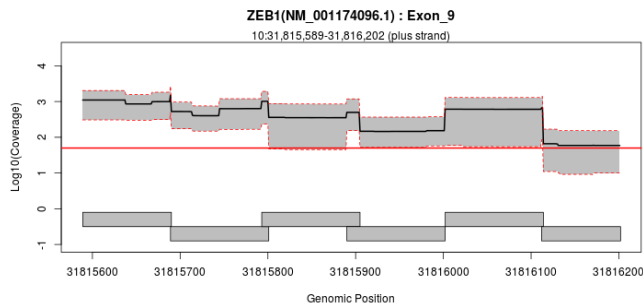
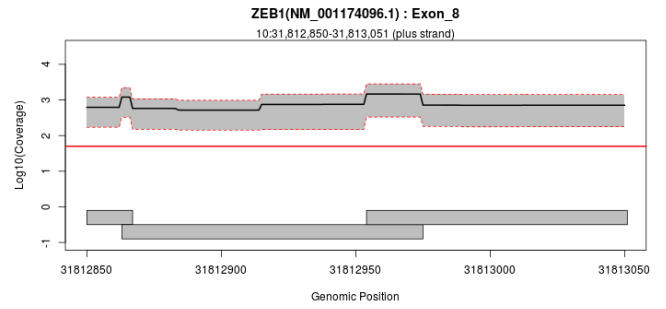
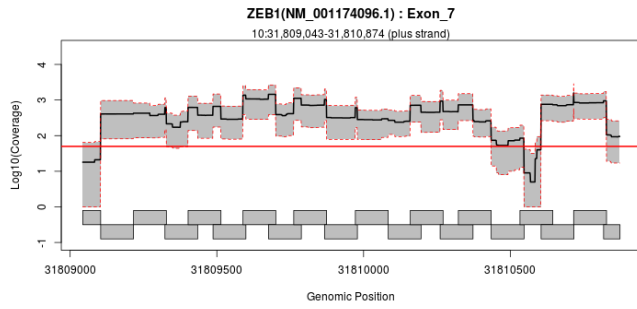
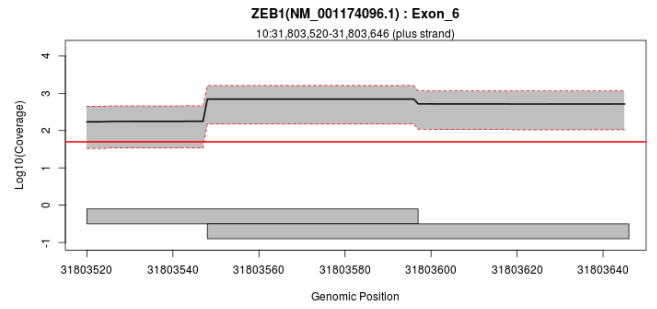
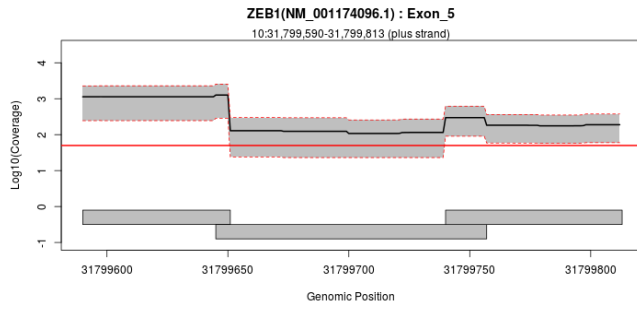
## Exon Overview: ZEB1 : NM\_001174095.1



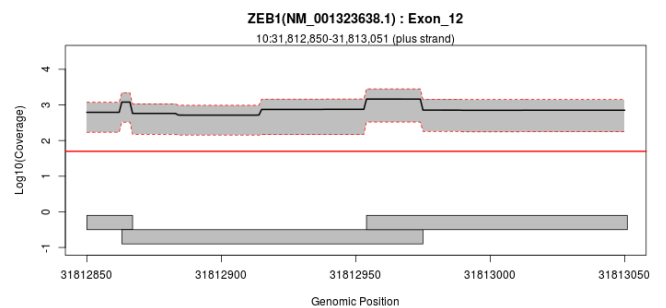
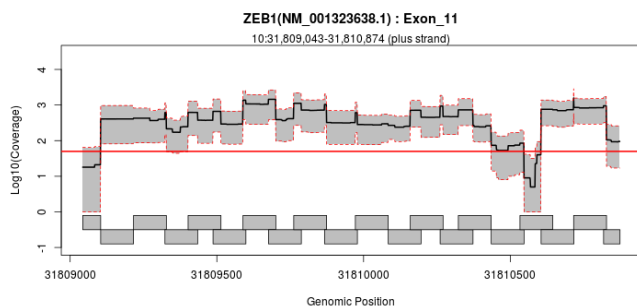
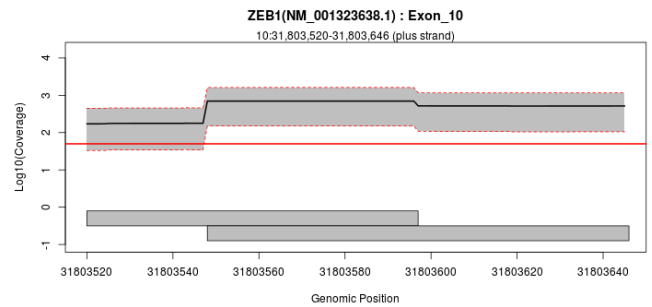
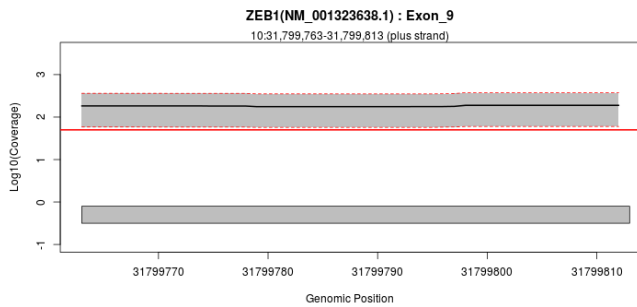


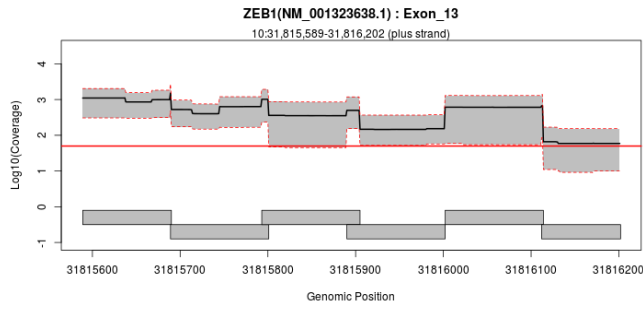
## Exon Overview: ZEB1 : NM\_001174096.1



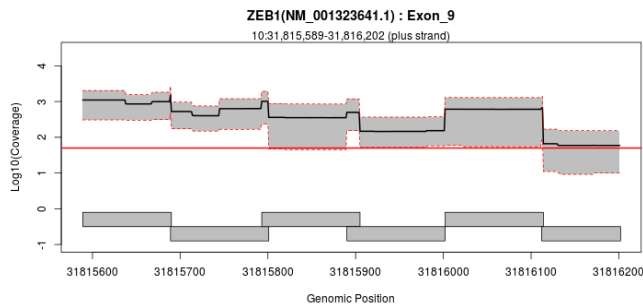
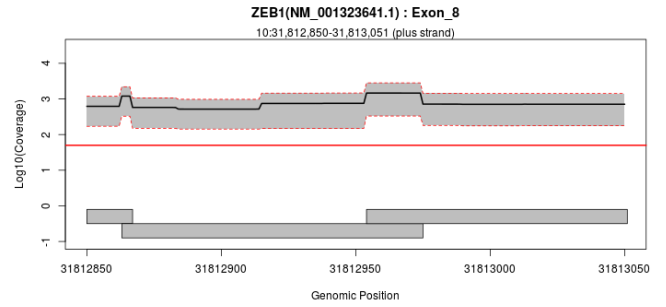
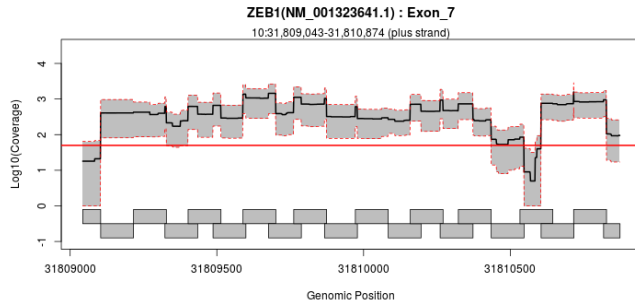
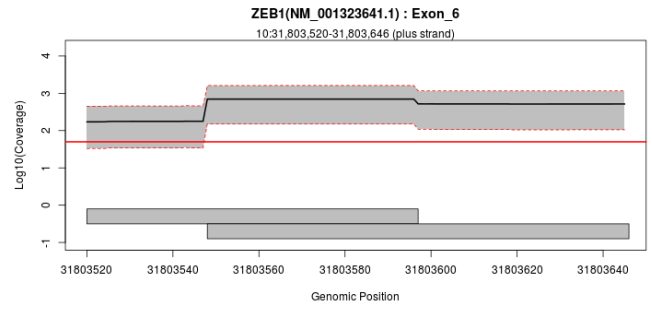
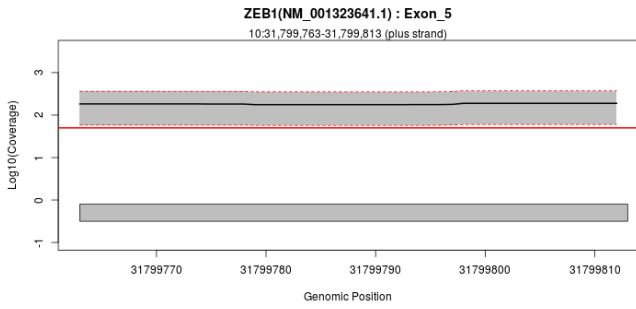


## Exon Overview: ZEB1 : NM\_001323638.1

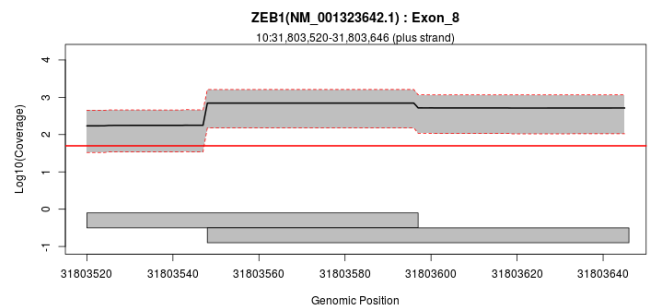
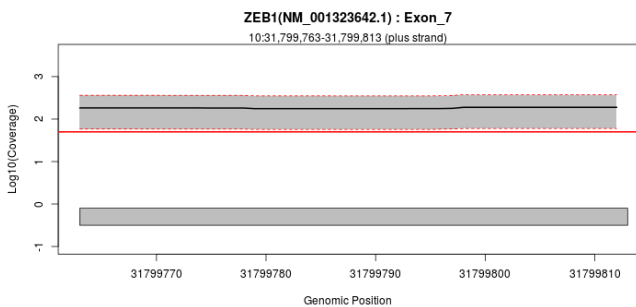


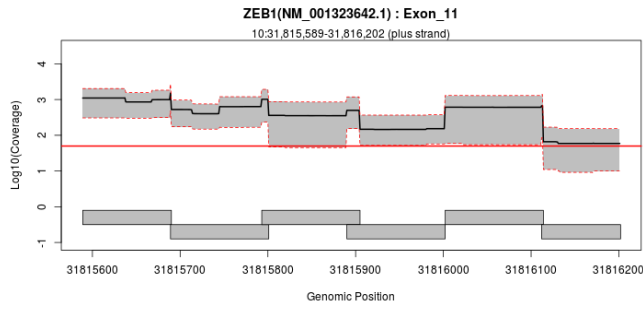
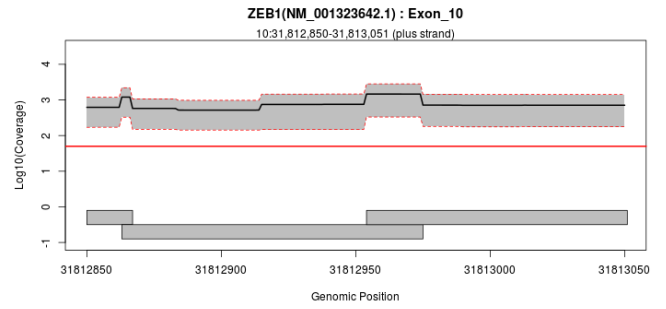
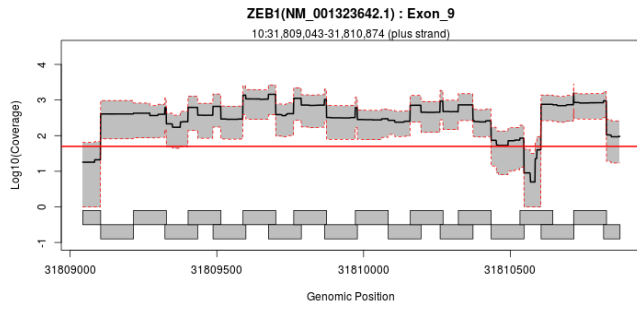


**Exon Overview: ZEB1 : NM\_001323641.1**

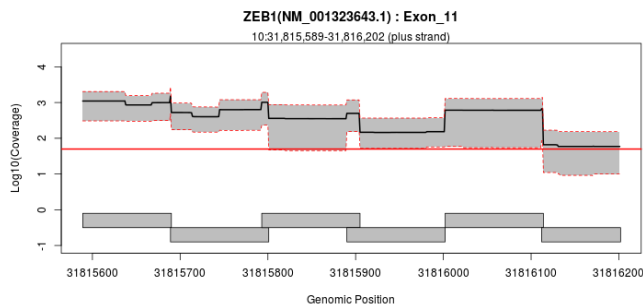
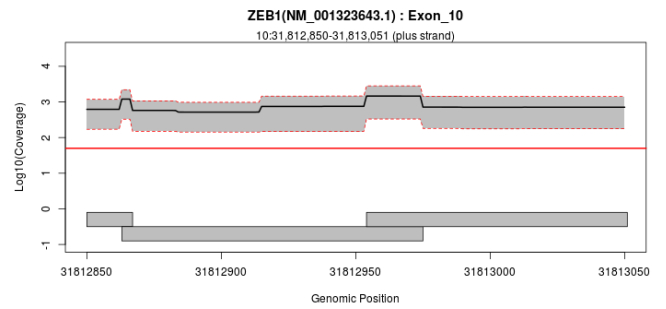
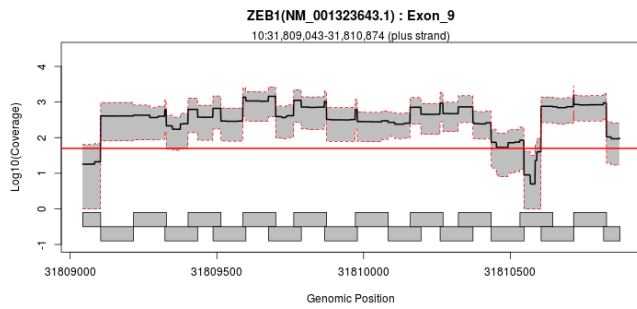
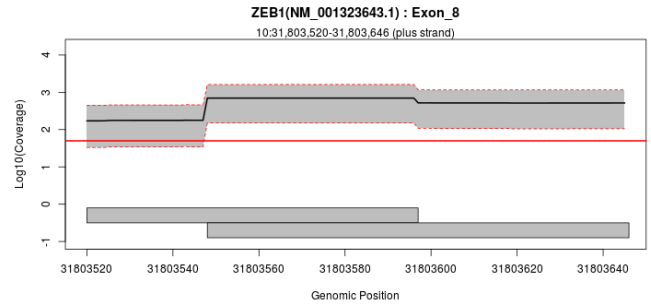
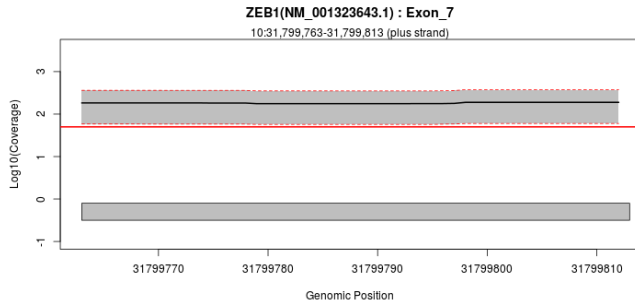


**Exon Overview: ZEB1 : NM\_001323642.1**

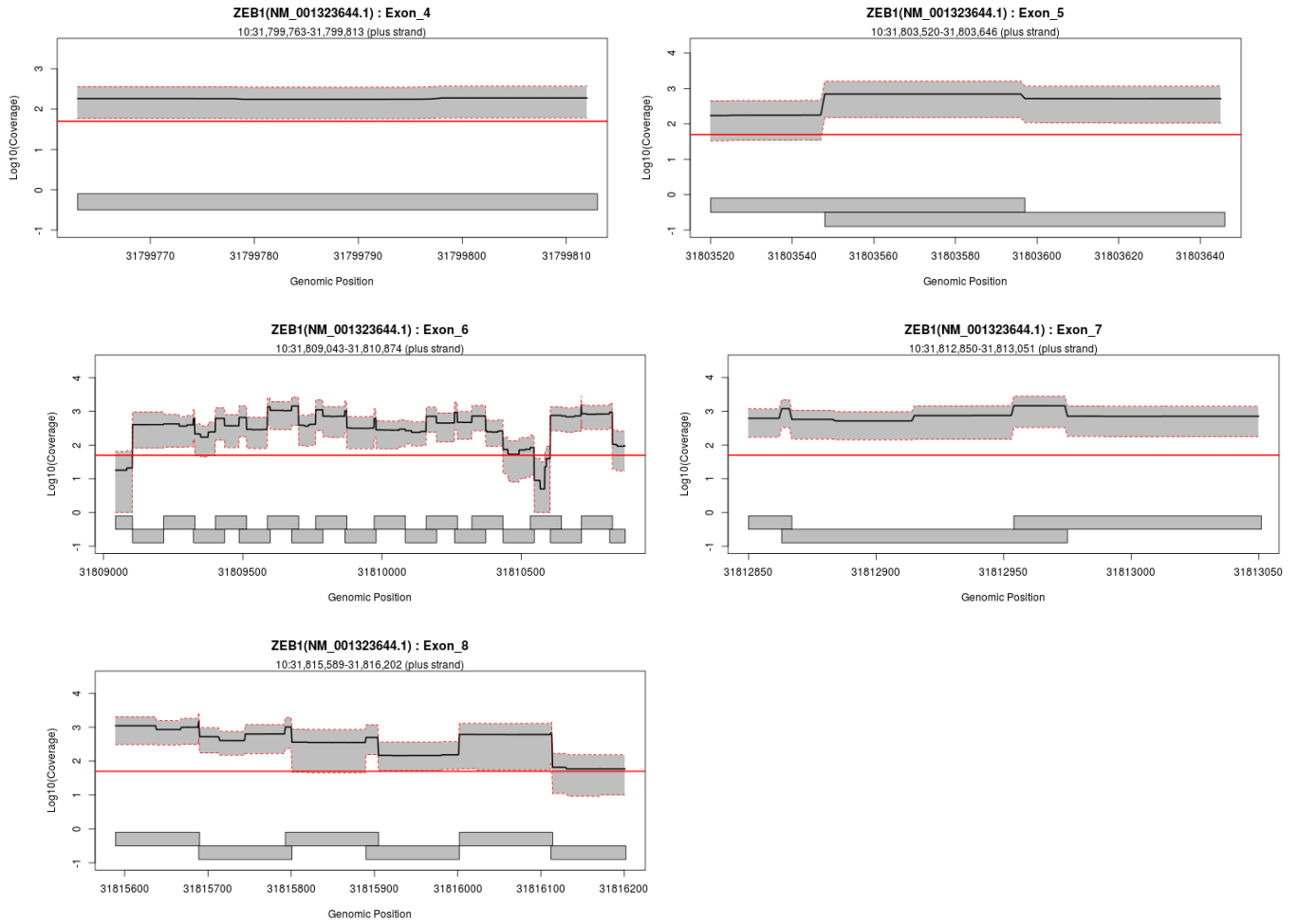




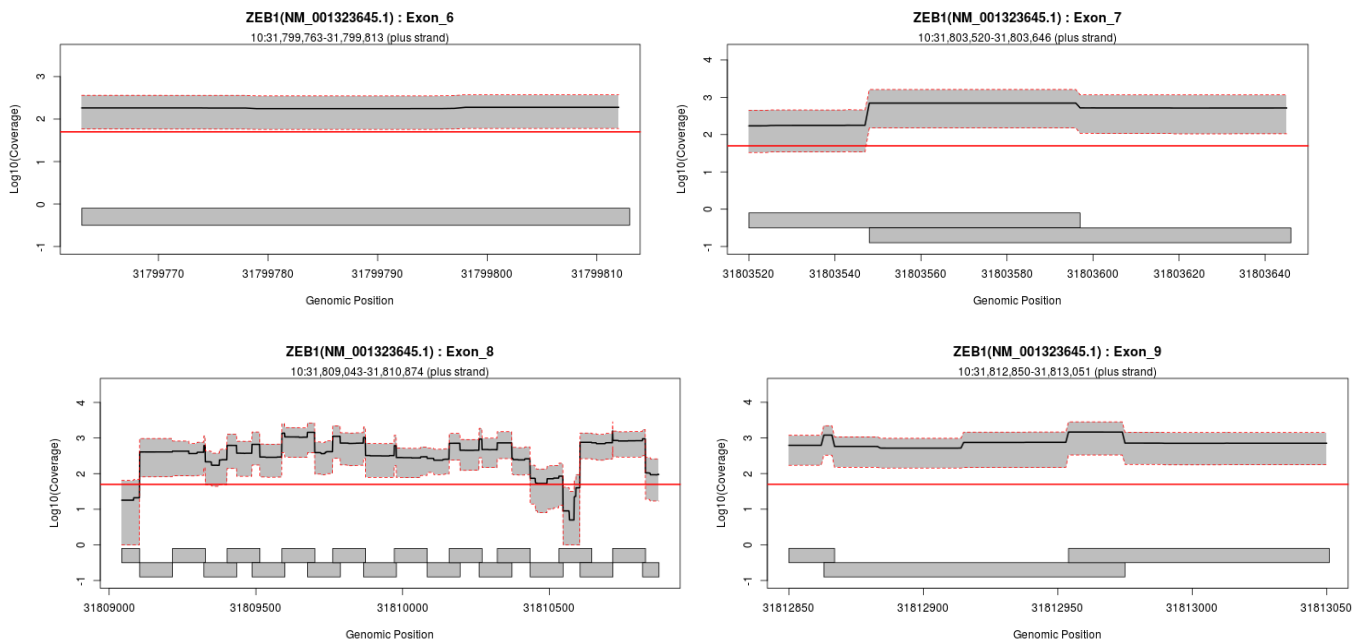
### Exon Overview: ZEB1 : NM\_001323643.1

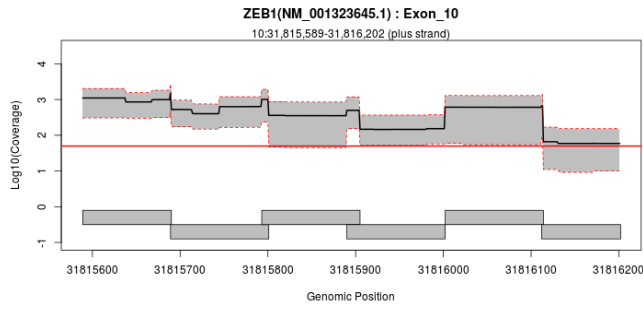


## Exon Overview: ZEB1 : NM\_001323644.1

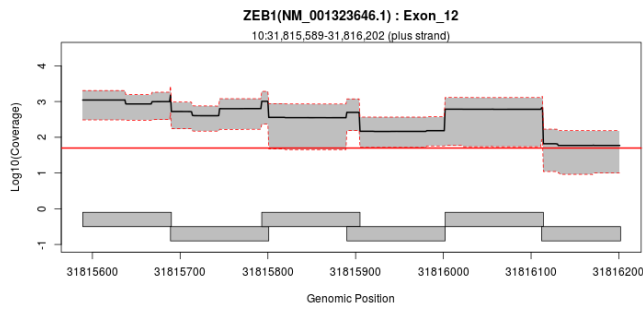
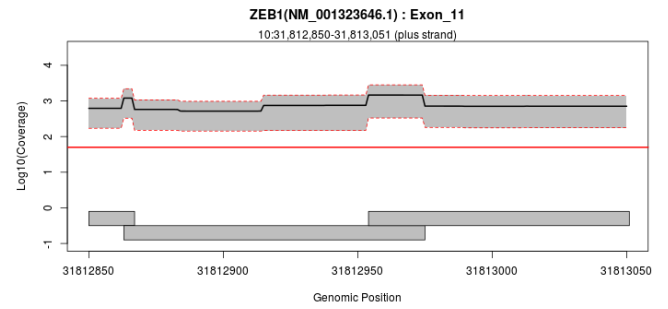
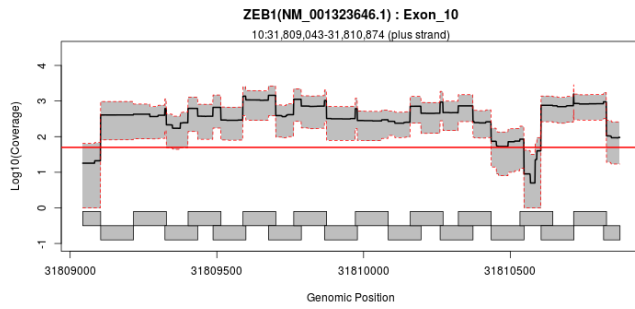
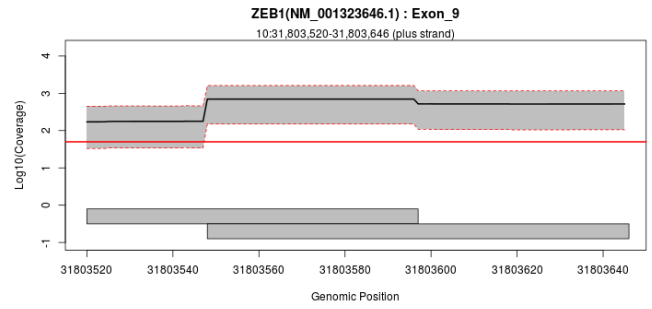
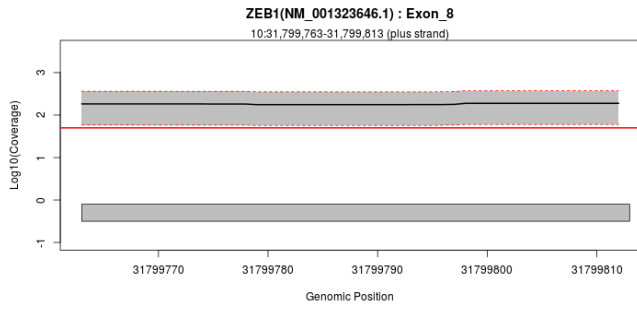


## Exon Overview: ZEB1 : NM\_001323645.1

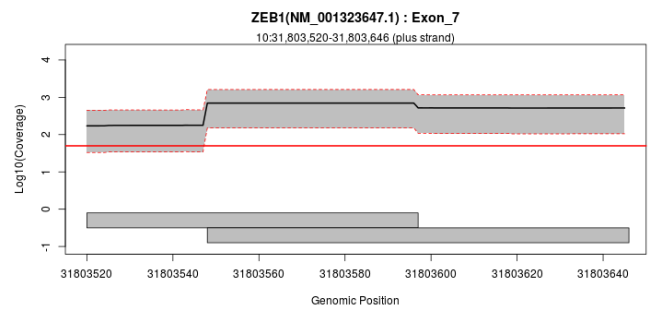
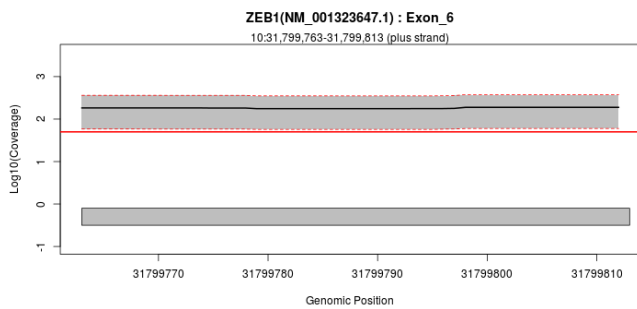




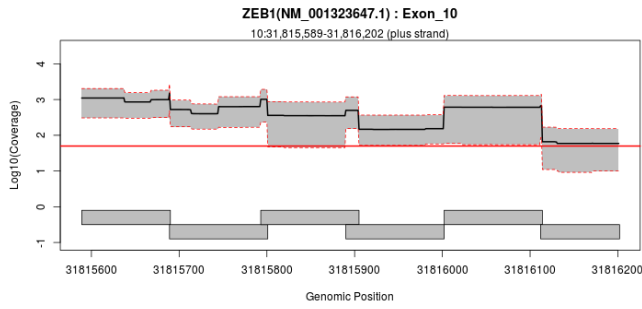
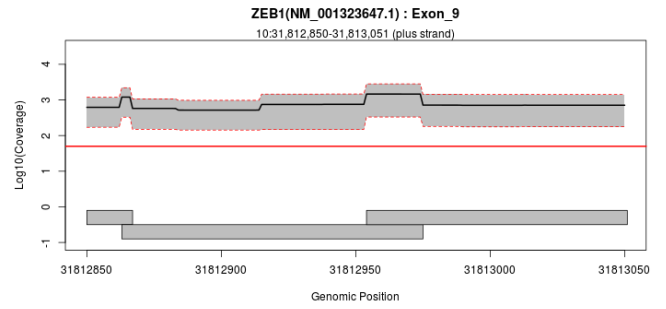
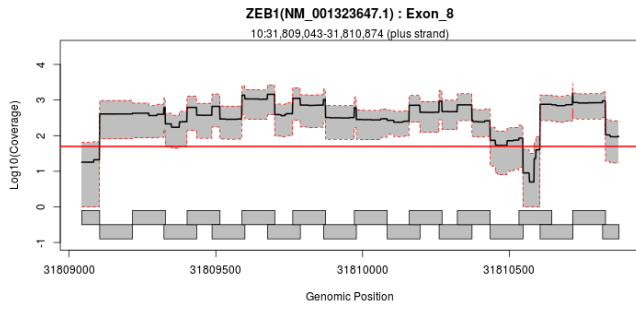
## Exon Overview: ZEB1 : NM\_001323646.1



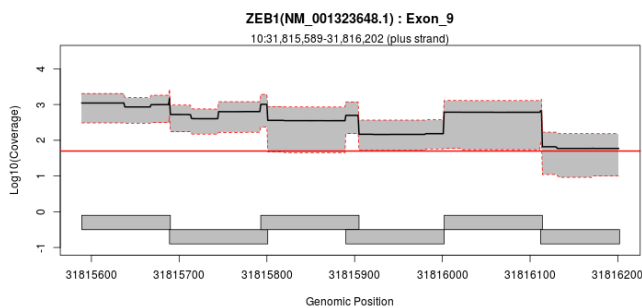
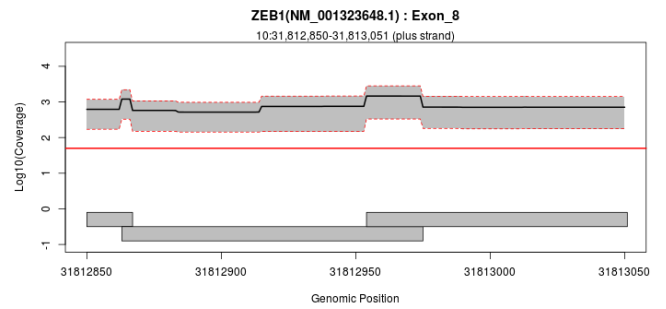
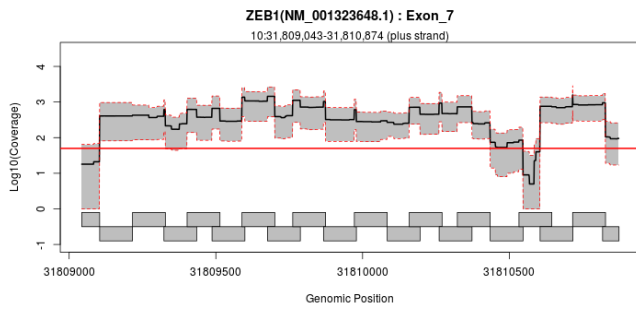
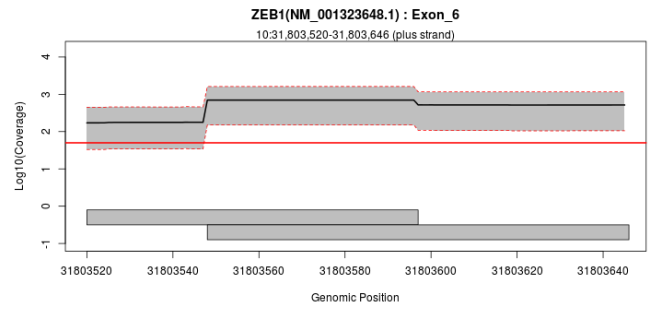
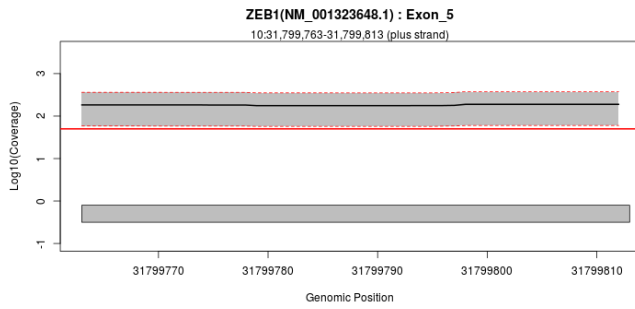
## Exon Overview: ZEB1 : NM\_001323647.1



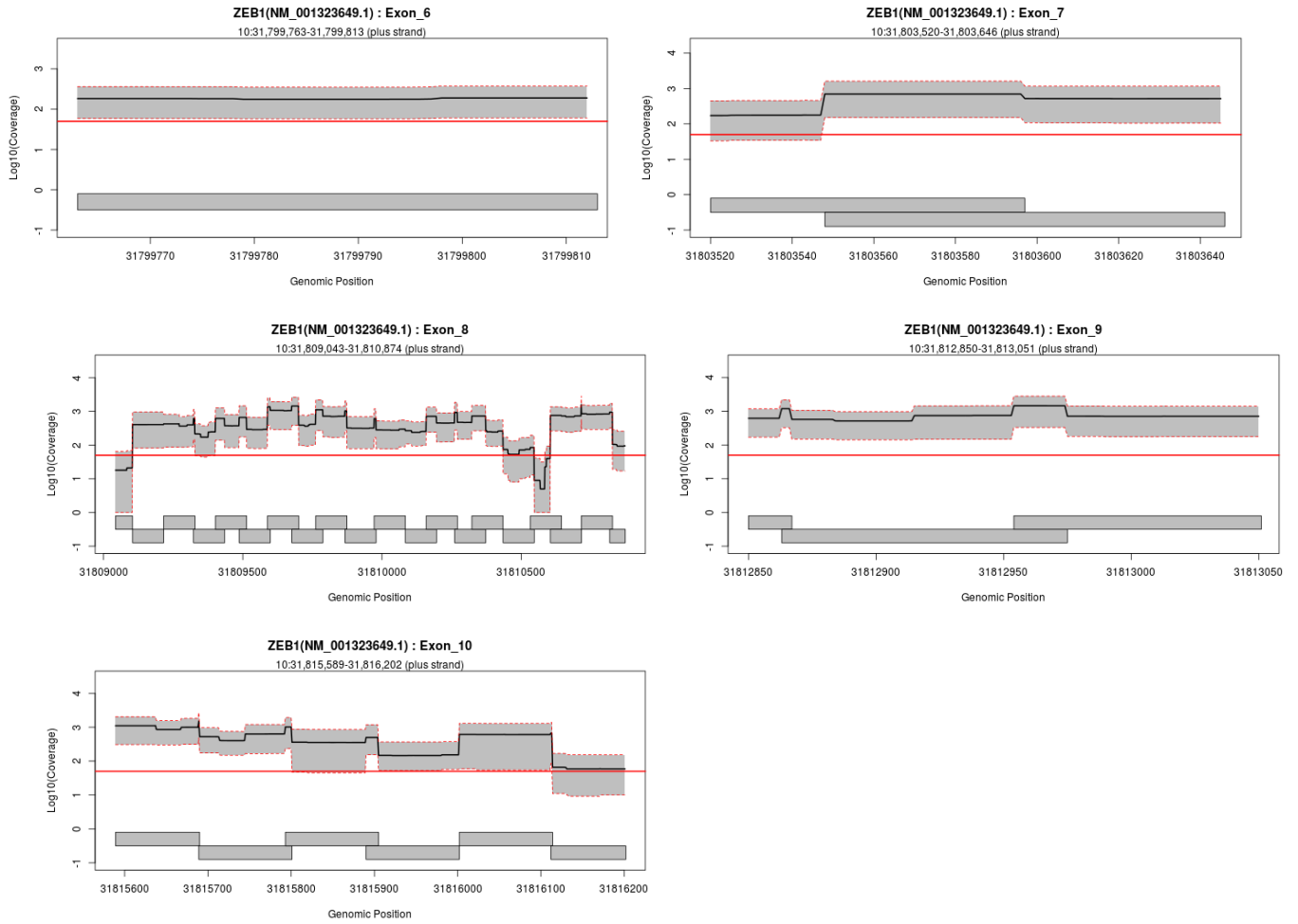




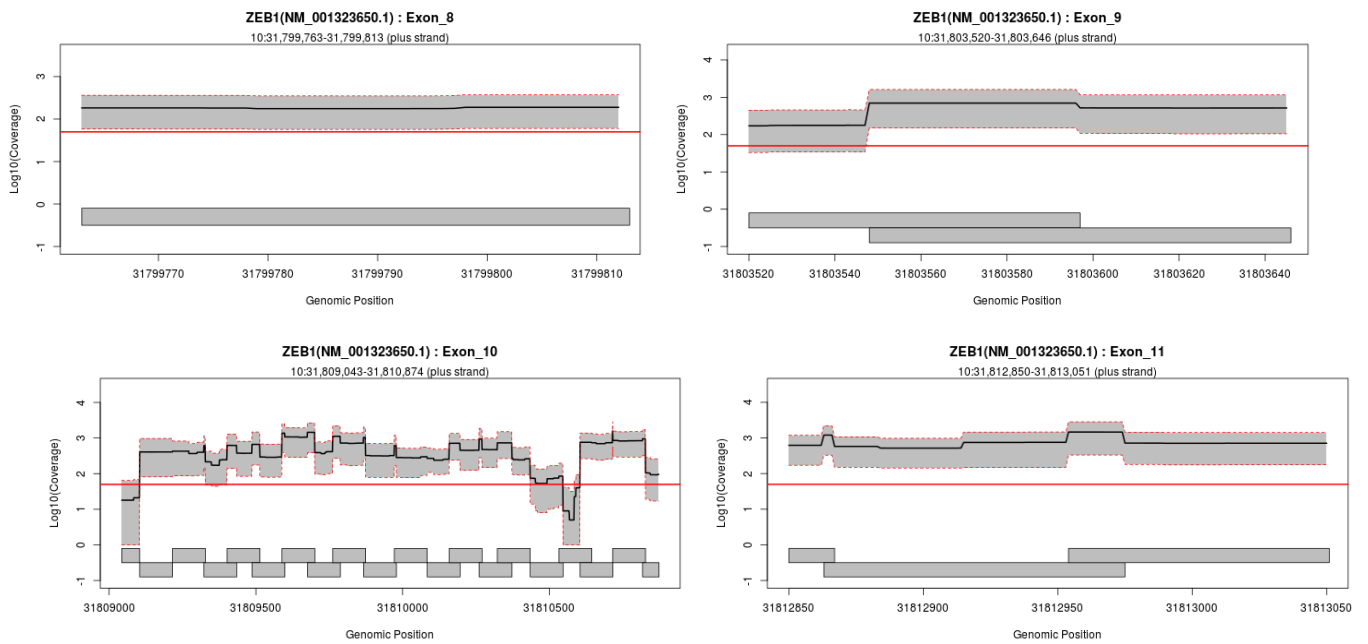
## Exon Overview: ZEB1 : NM\_001323648.1

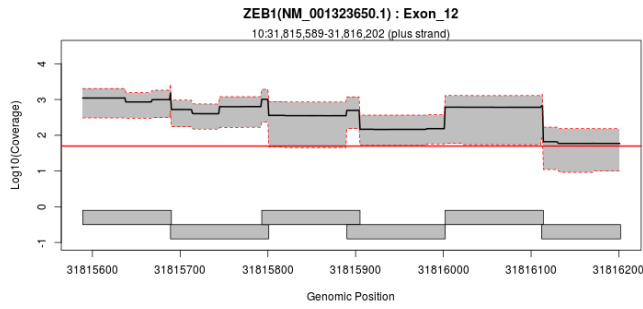


## Exon Overview: ZEB1 : NM\_001323649.1

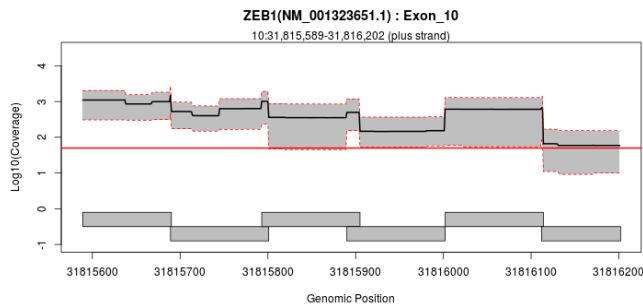
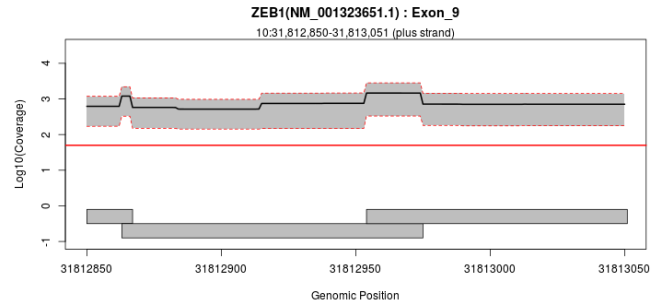
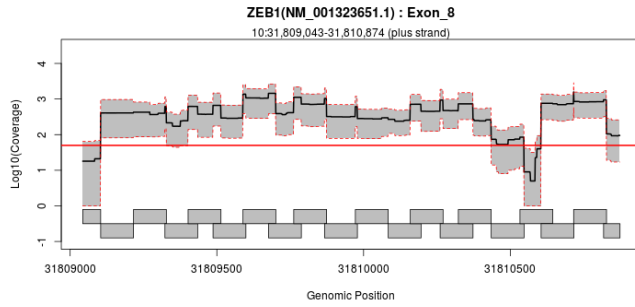
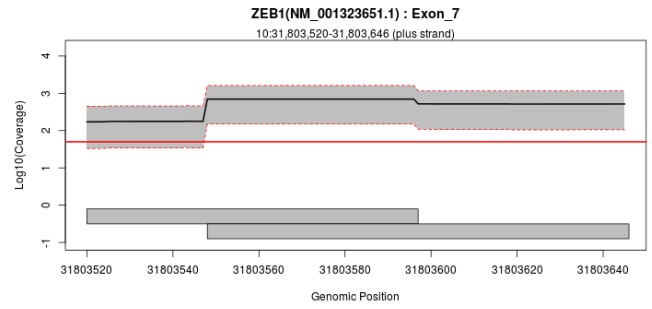
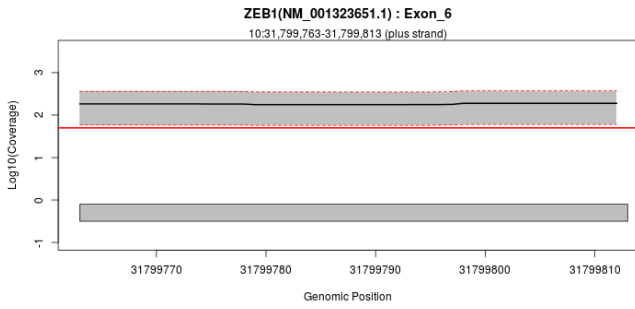


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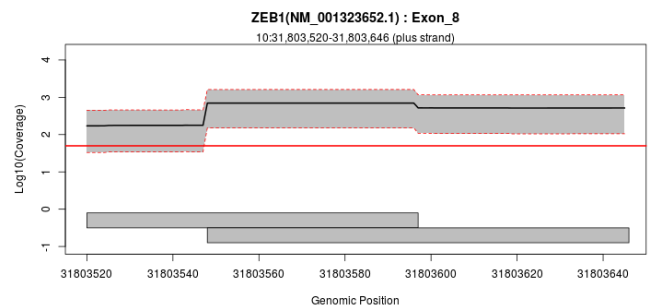
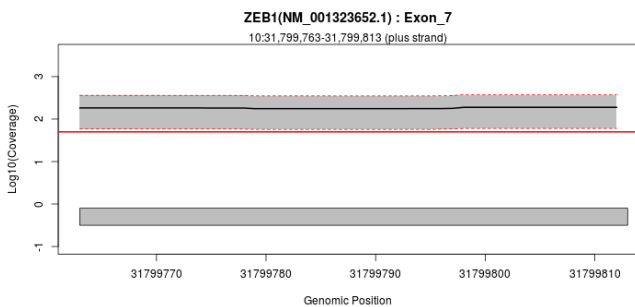


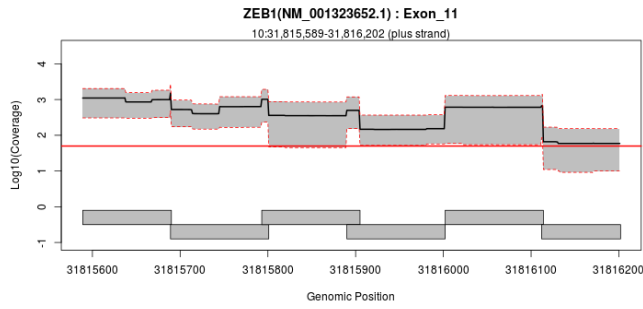
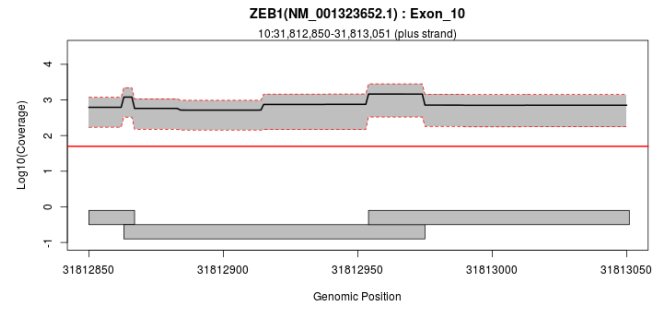
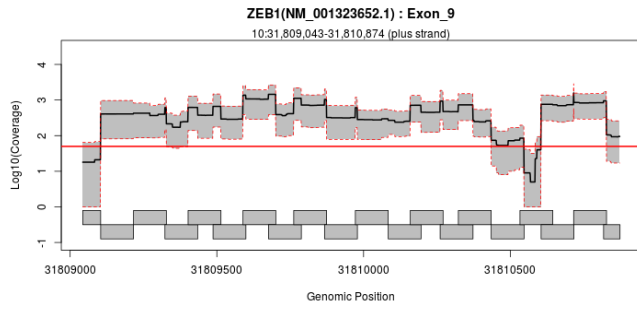


## Exon Overview: ZEB1 : NM\_001323651.1

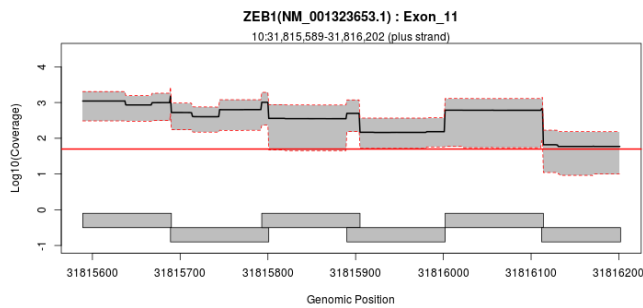
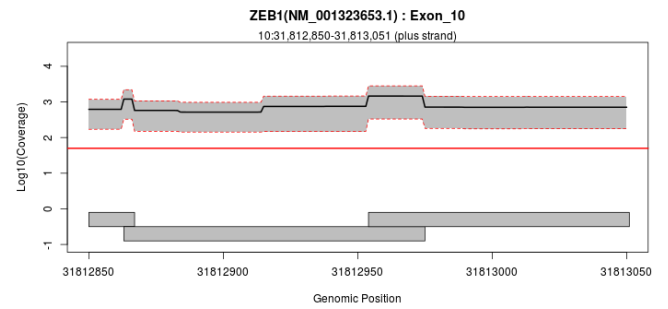
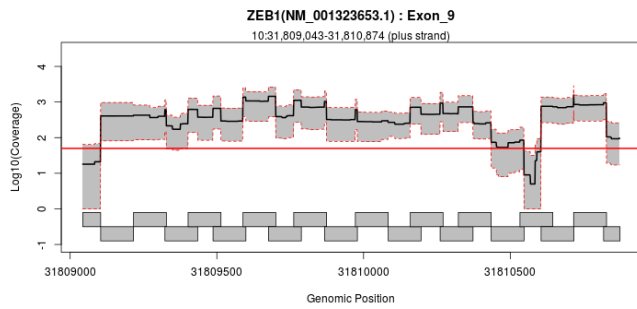
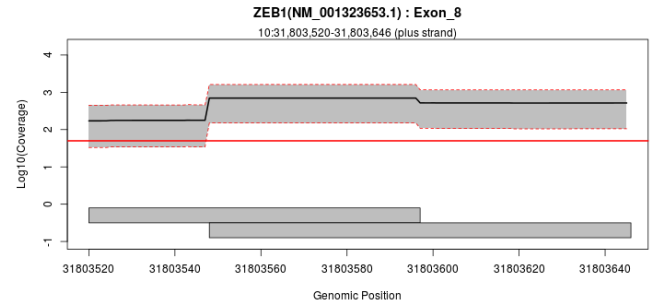
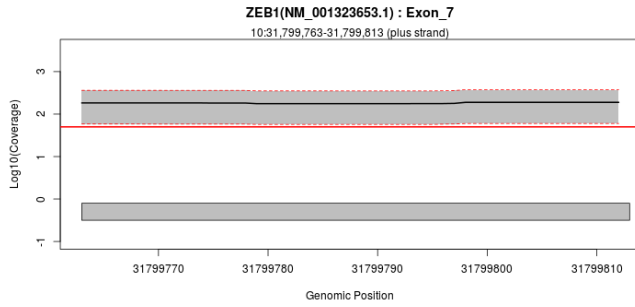


## Exon Overview: ZEB1 : NM\_001323652.1

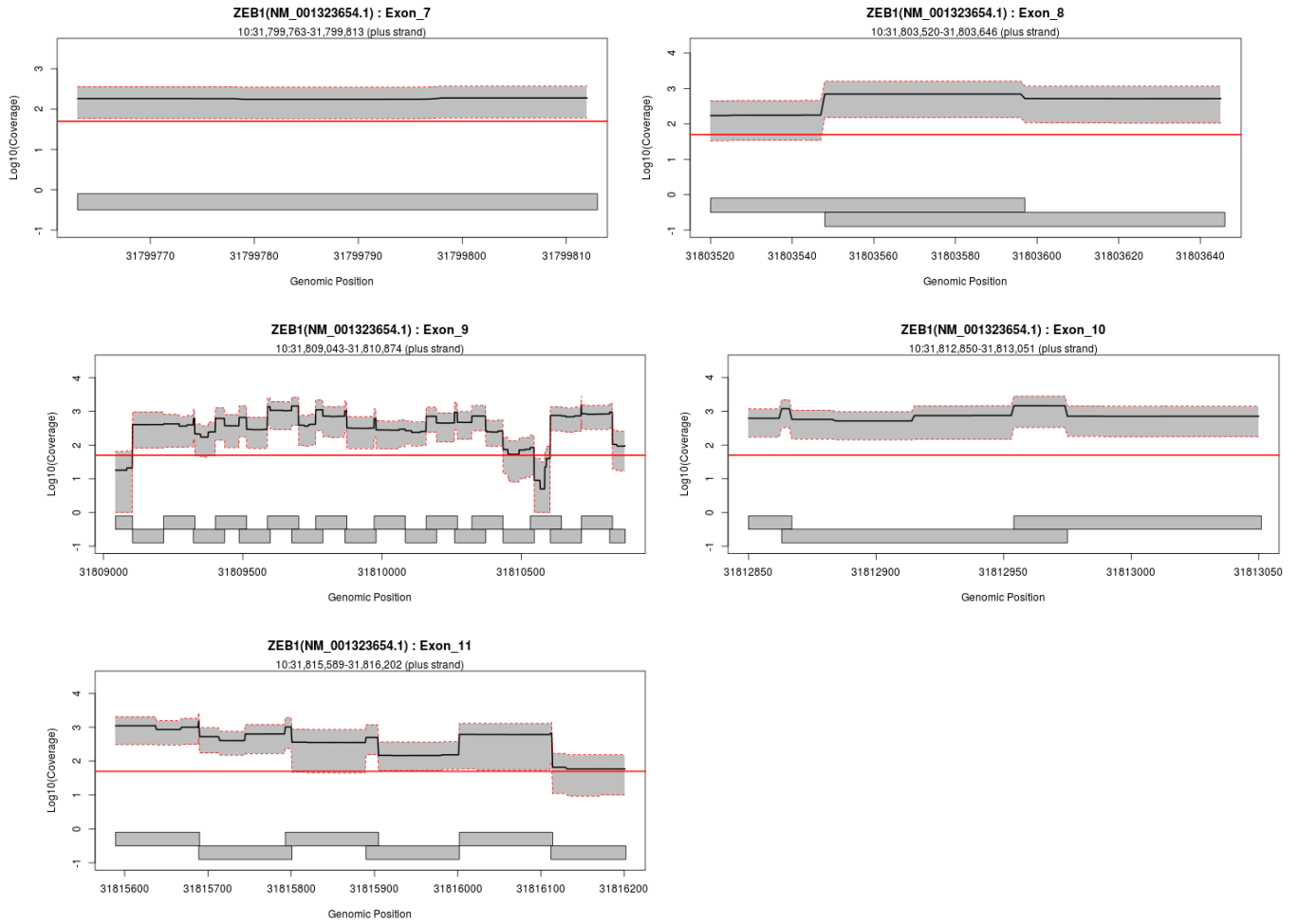




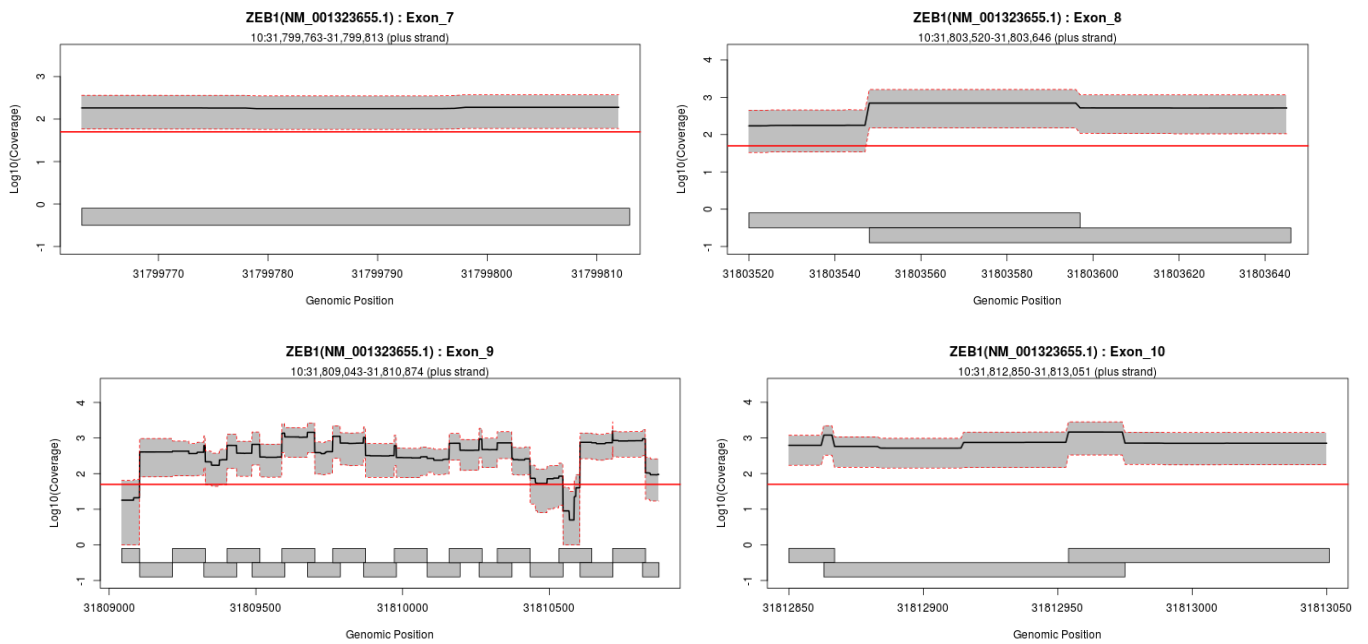
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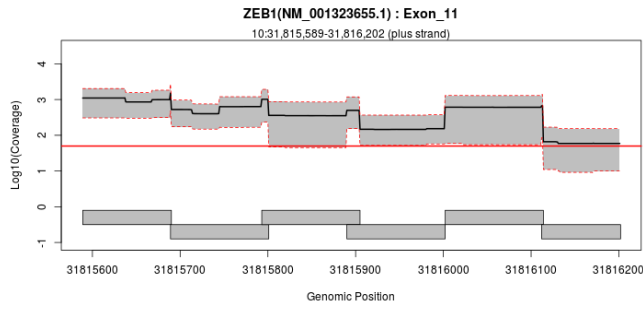


## Exon Overview: ZEB1 : NM\_001323654.1

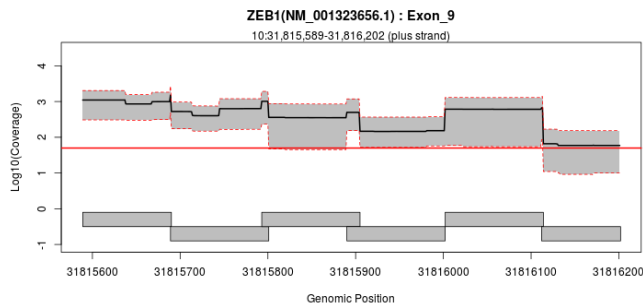
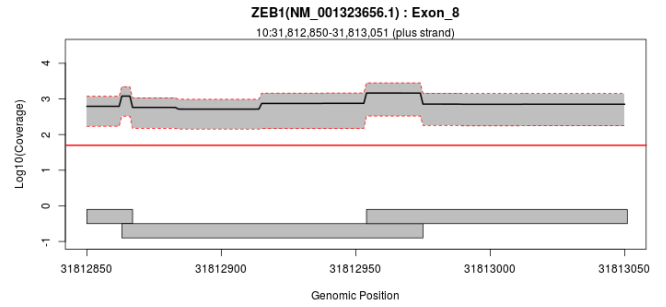
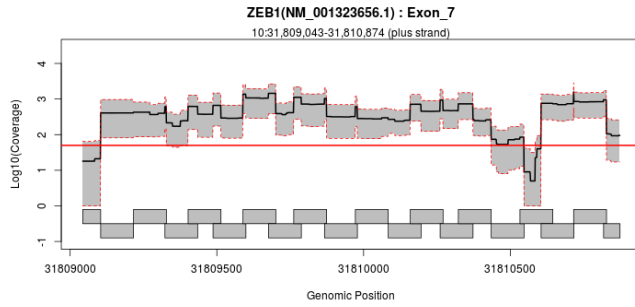
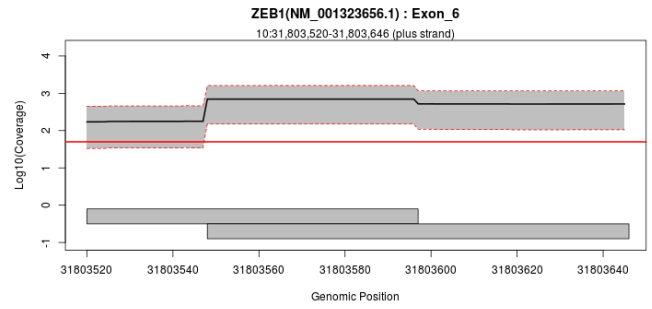
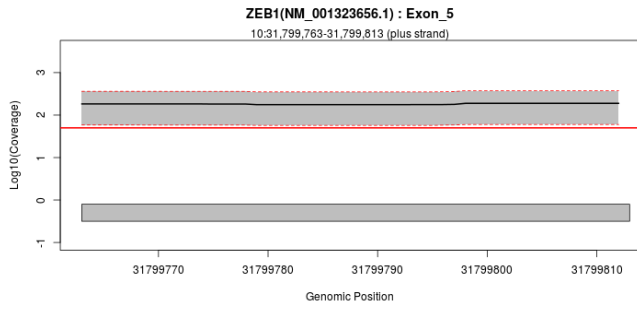


## Exon Overview: ZEB1 : NM\_001323655.1

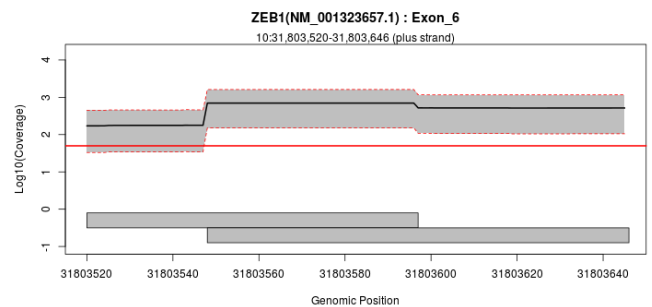
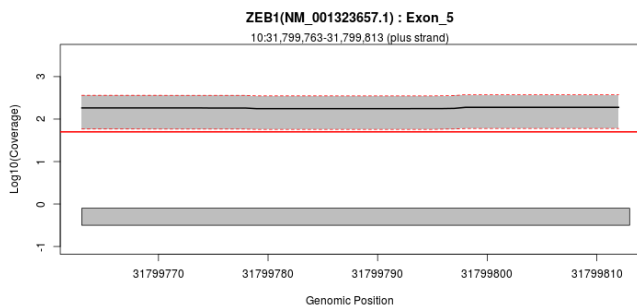


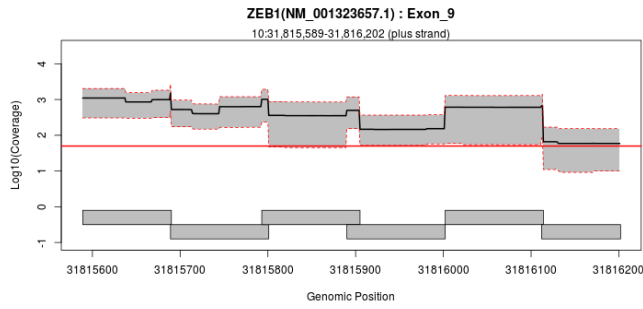
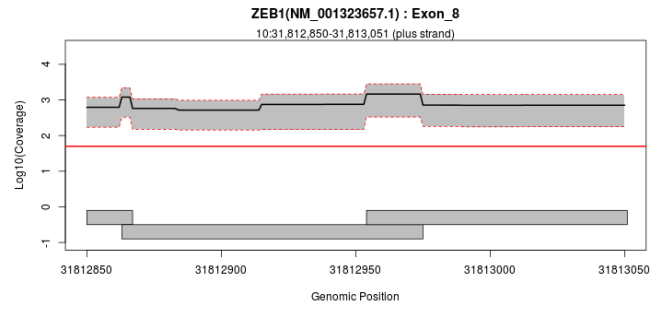
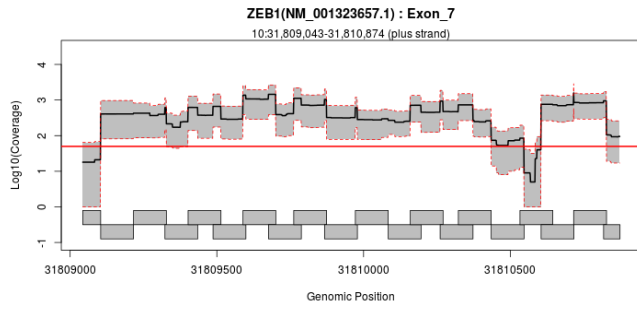


## Exon Overview: ZEB1 : NM\_001323656.1

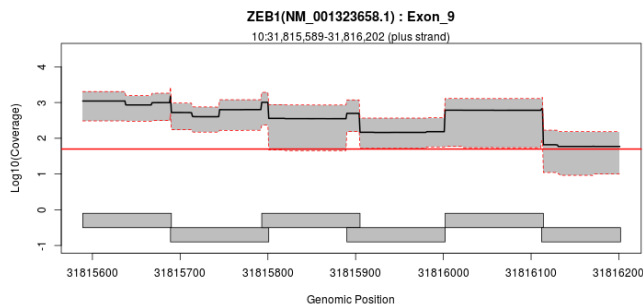
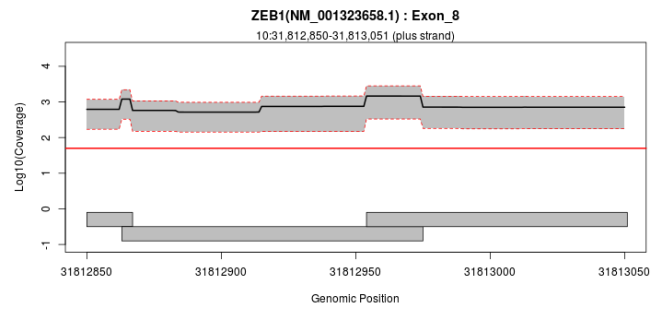
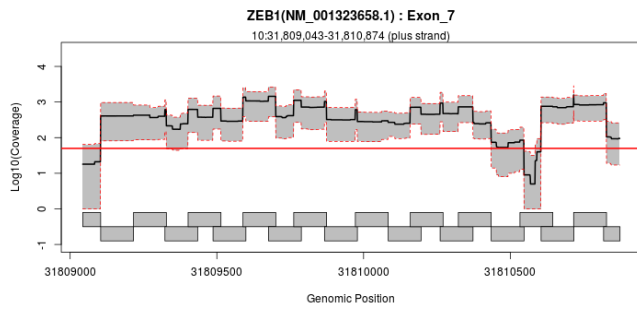
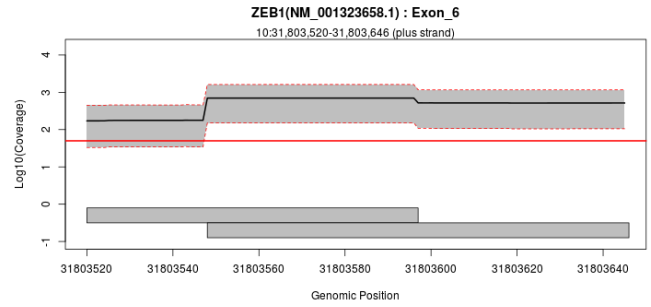
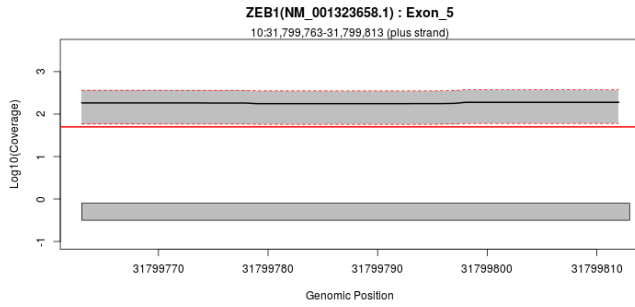


## Exon Overview: ZEB1 : NM\_001323657.1

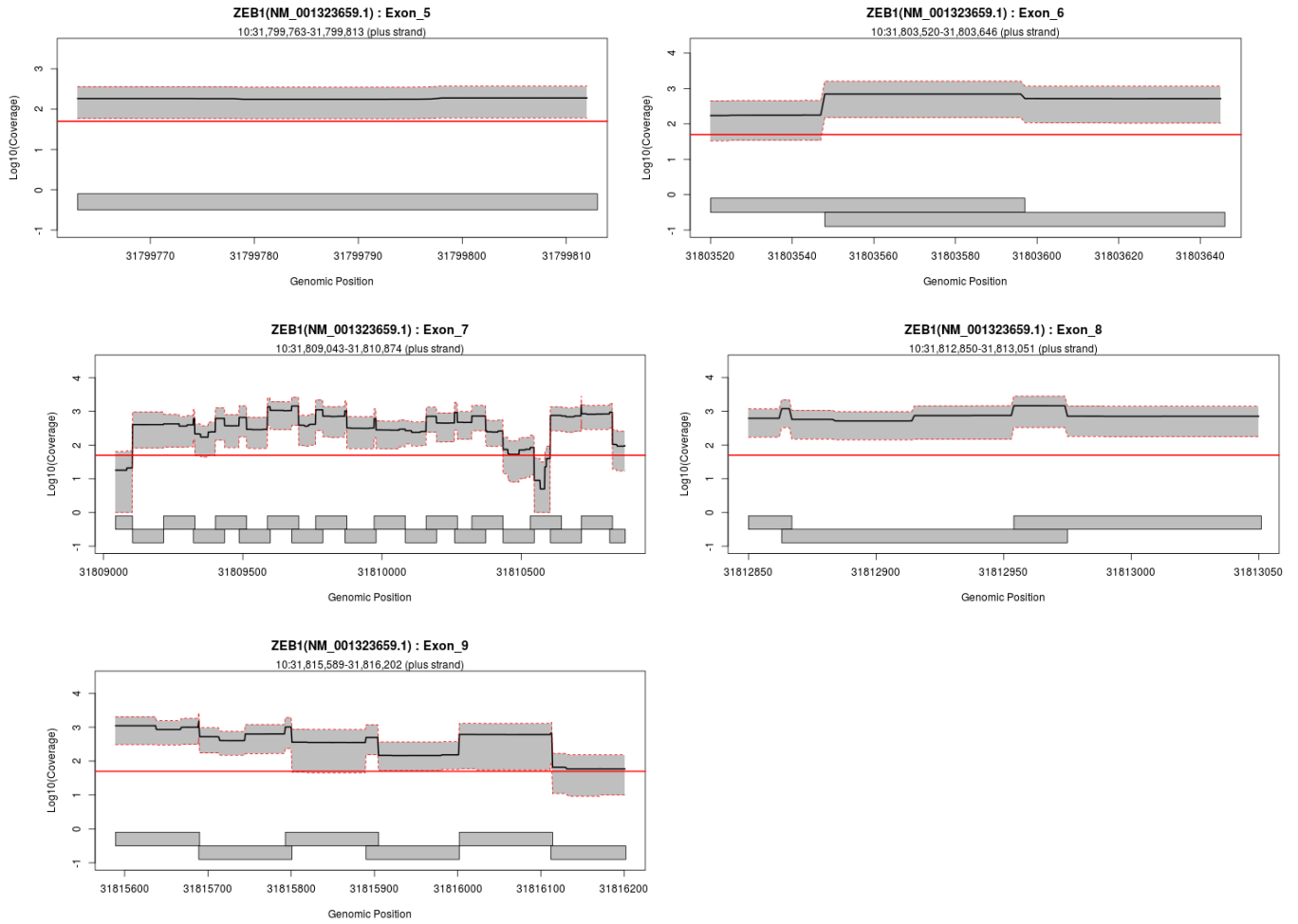




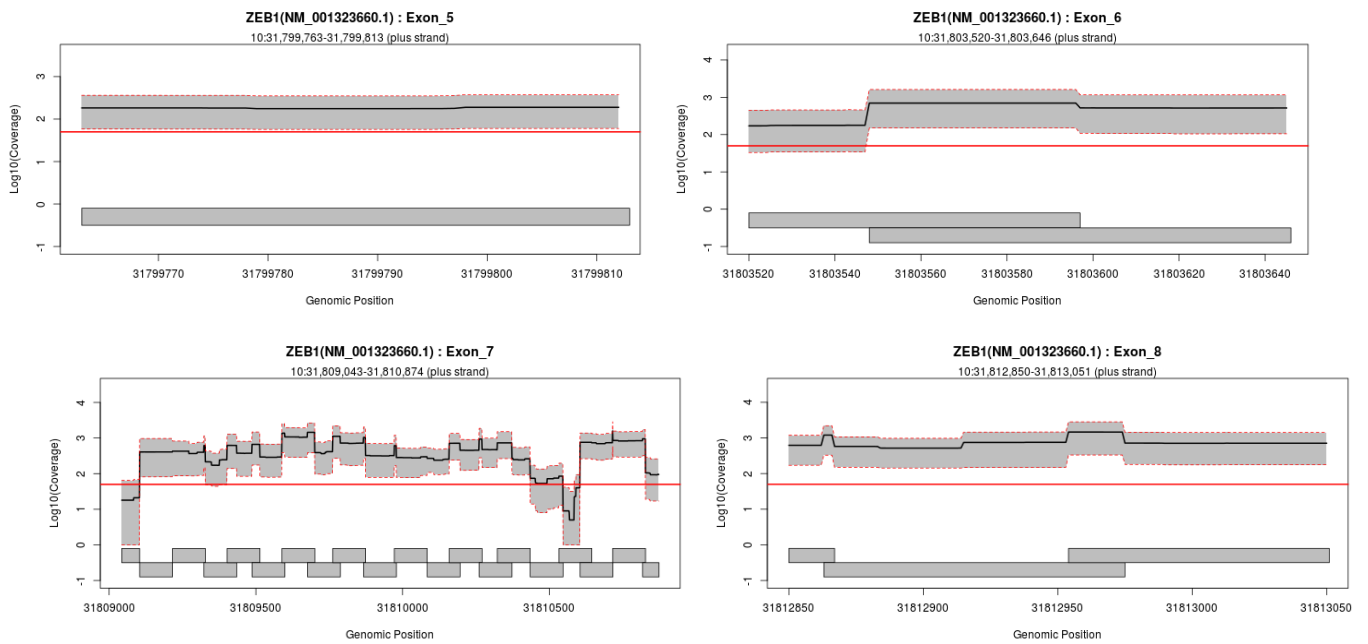
### Exon Overview: ZEB1 : NM\_001323658.1



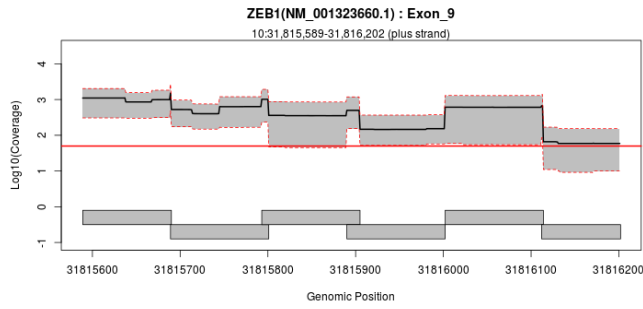
## Exon Overview: ZEB1 : NM\_001323659.1



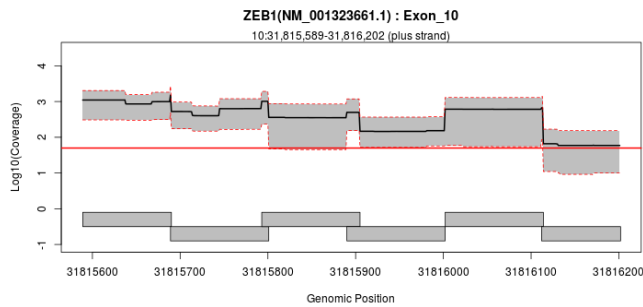
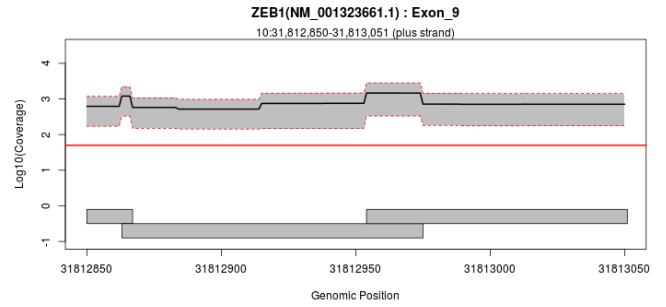
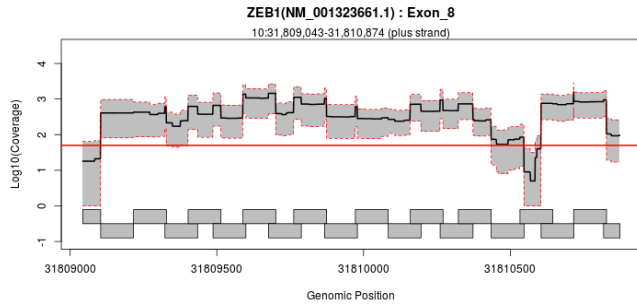
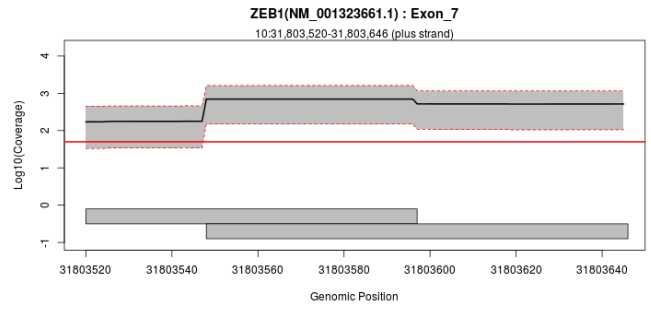
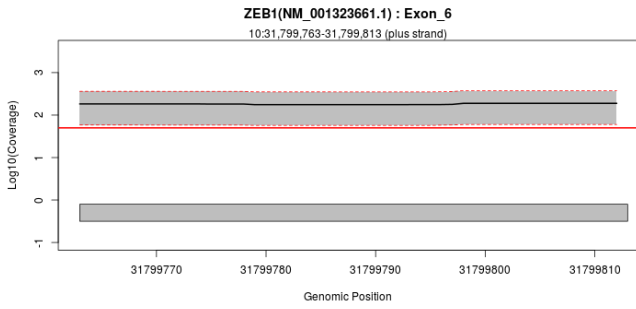
## Exon Overview: ZEB1 : NM\_001323660.1



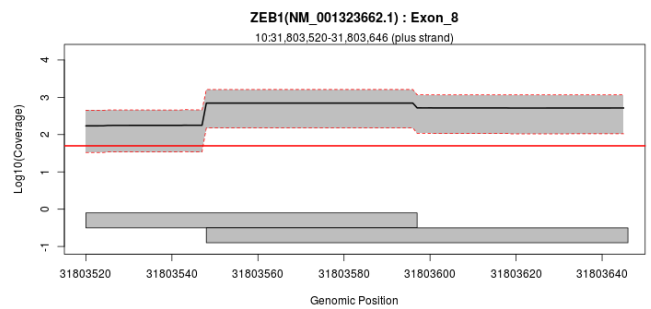
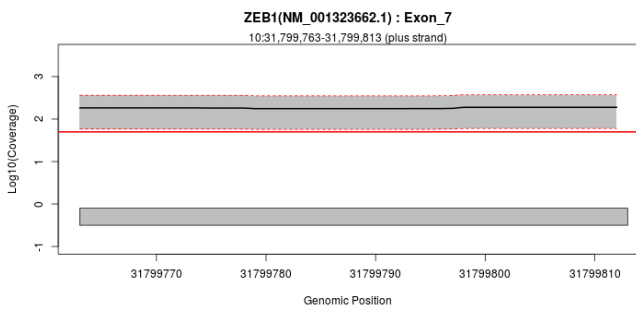


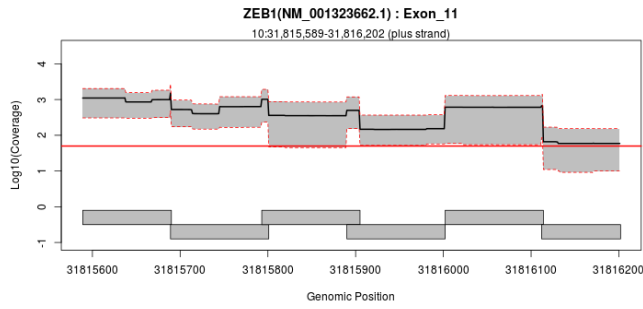
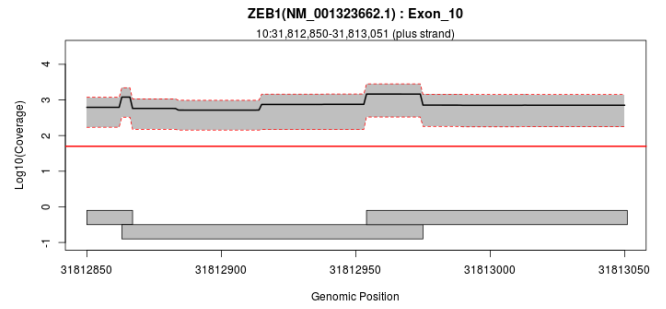
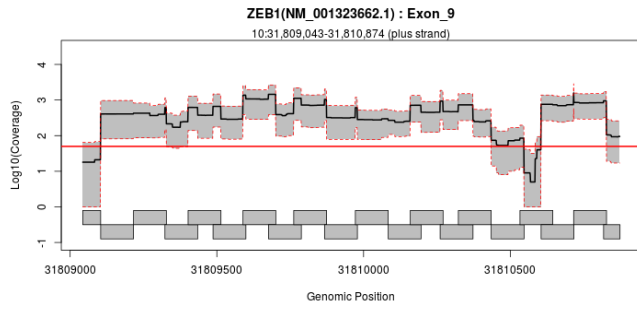


**Exon Overview: ZEB1 : NM\_001323661.1**

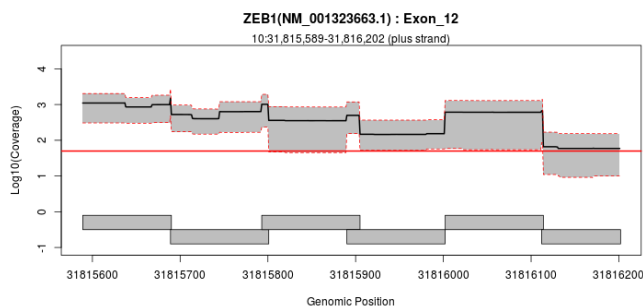
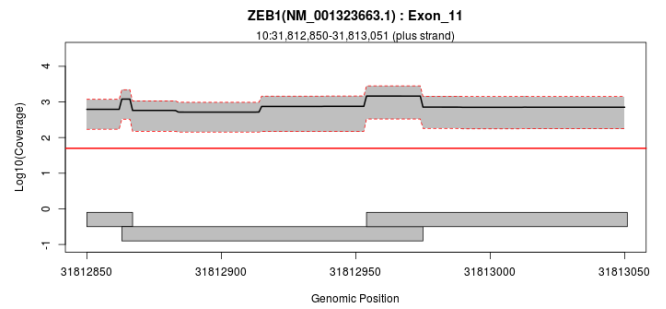
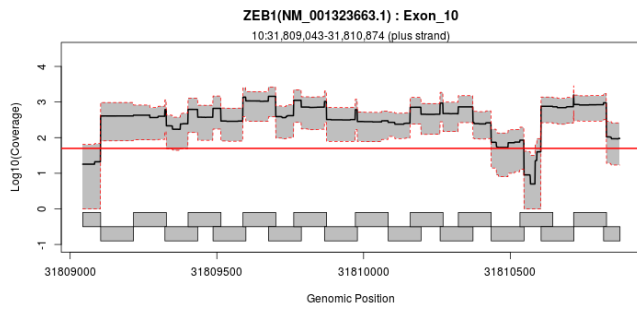
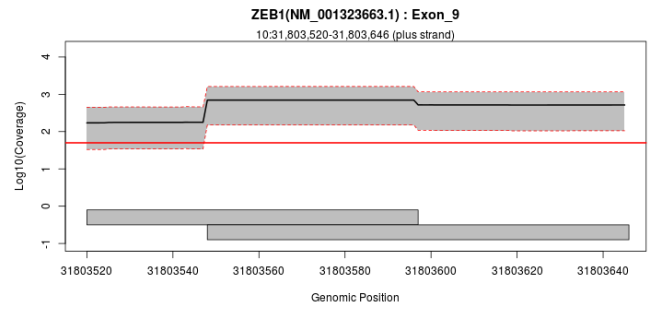
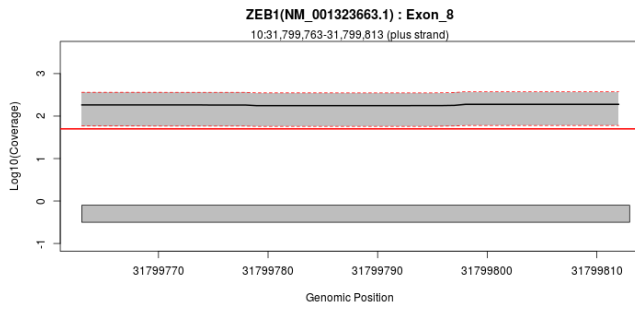


**Exon Overview: ZEB1 : NM\_001323662.1**

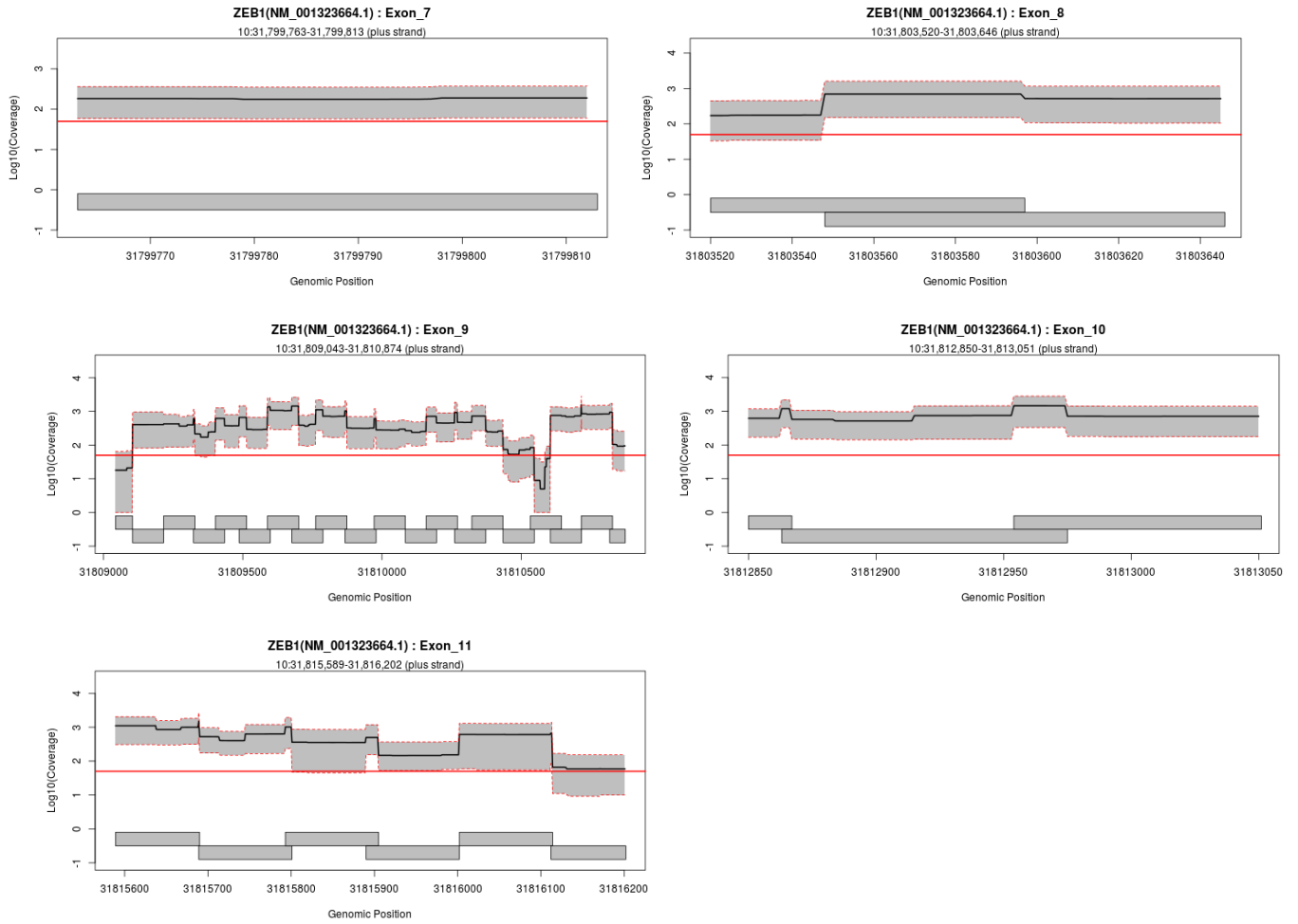




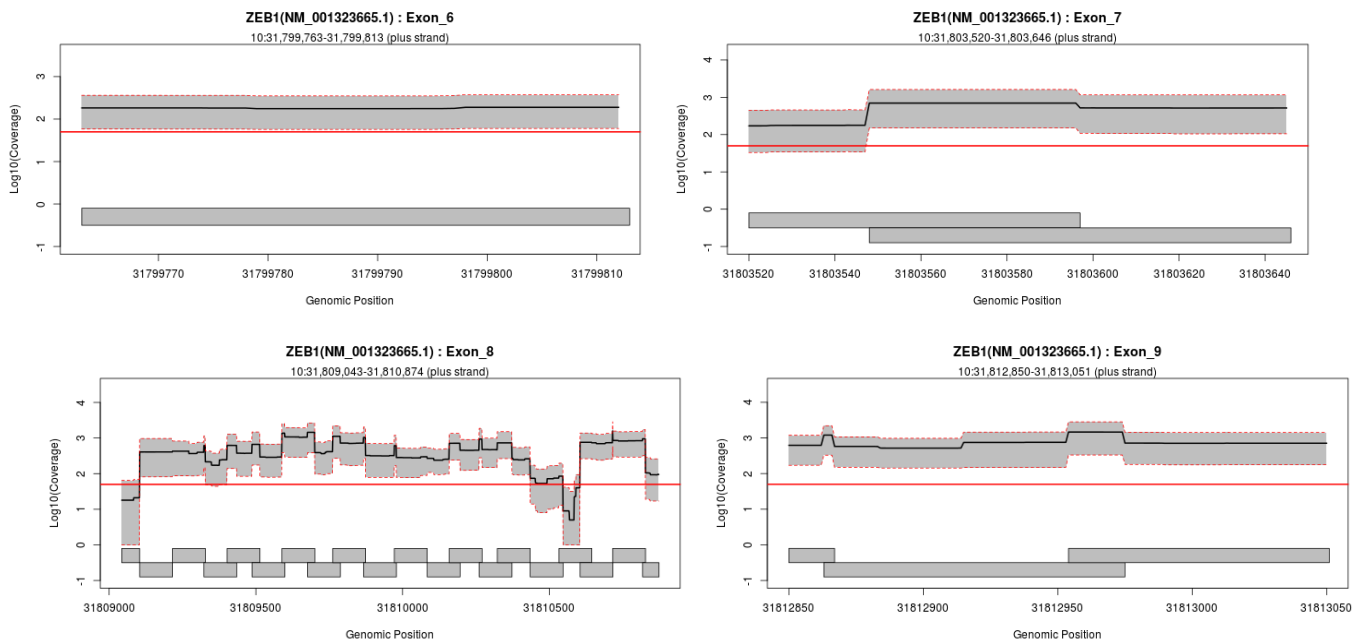
## Exon Overview: ZEB1 : NM\_001323663.1

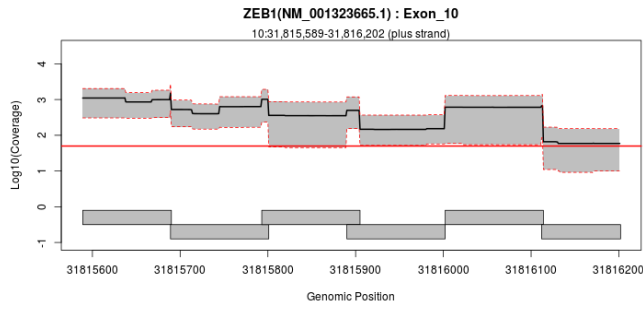


## Exon Overview: ZEB1 : NM\_001323664.1

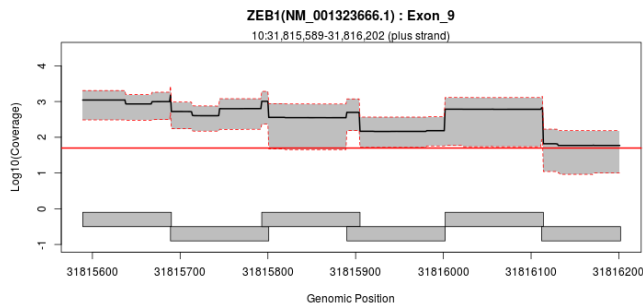
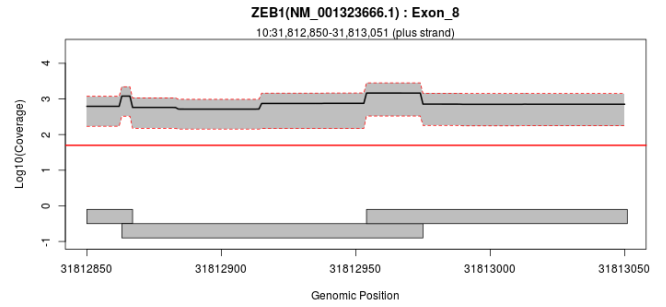
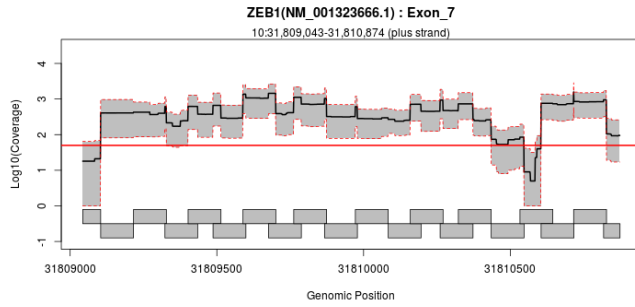
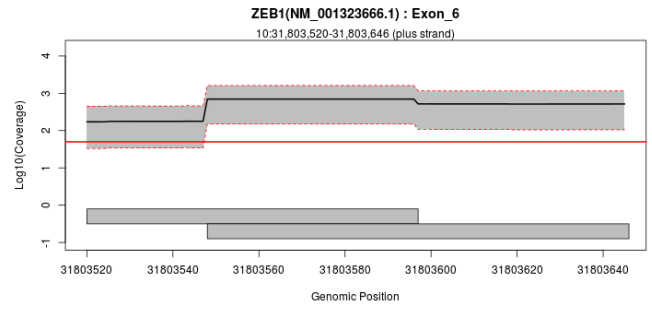
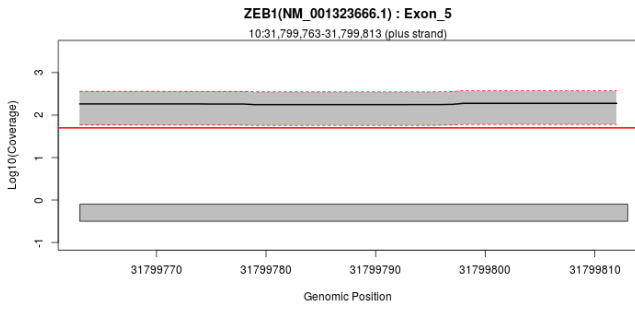


## Exon Overview: ZEB1 : NM\_001323665.1

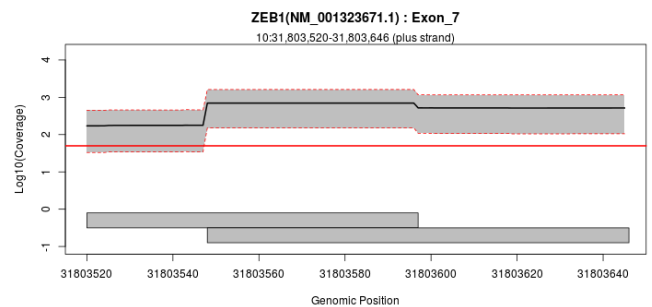
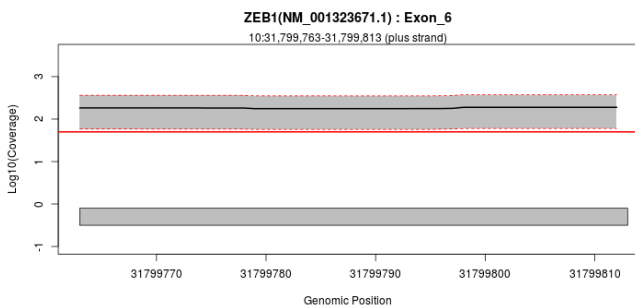


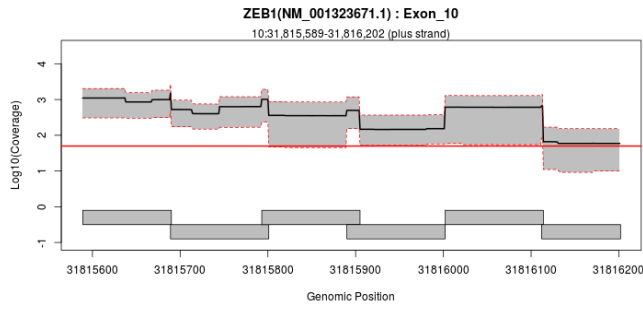
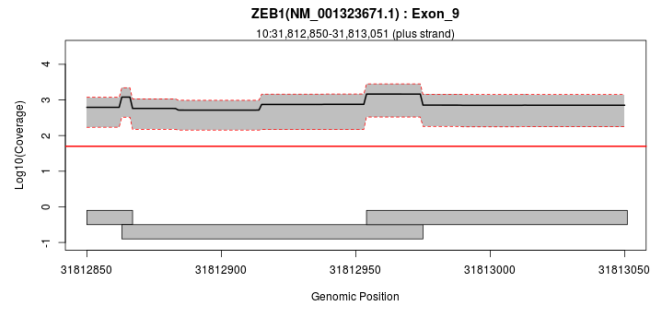
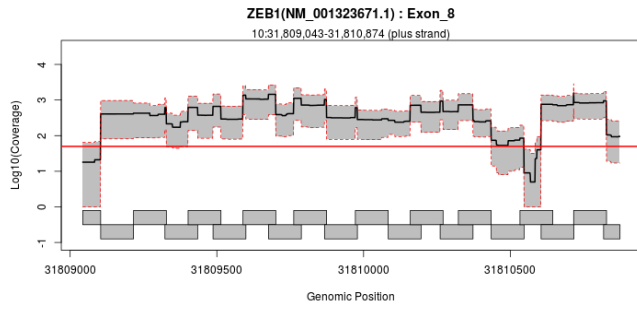


## Exon Overview: ZEB1 : NM\_001323666.1

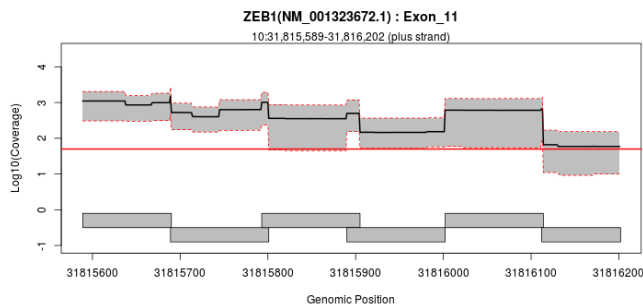
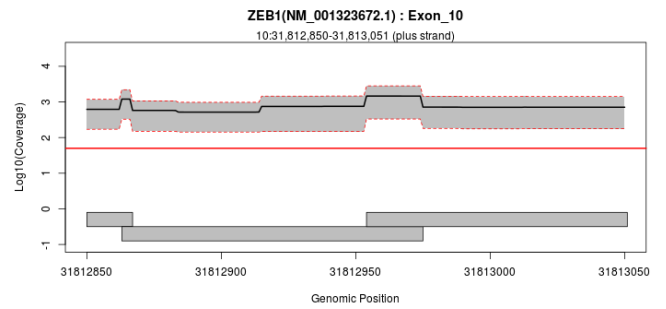
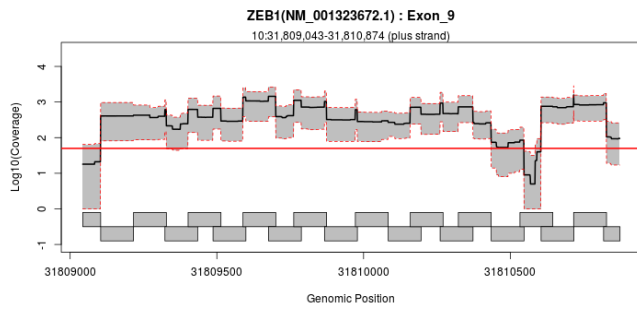
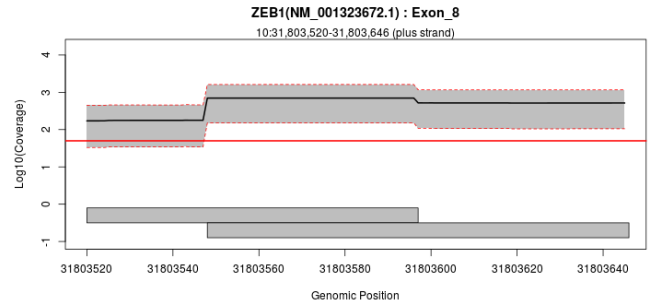
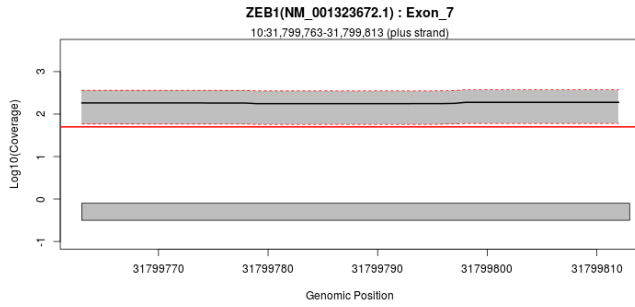


## Exon Overview: ZEB1 : NM\_001323671.1

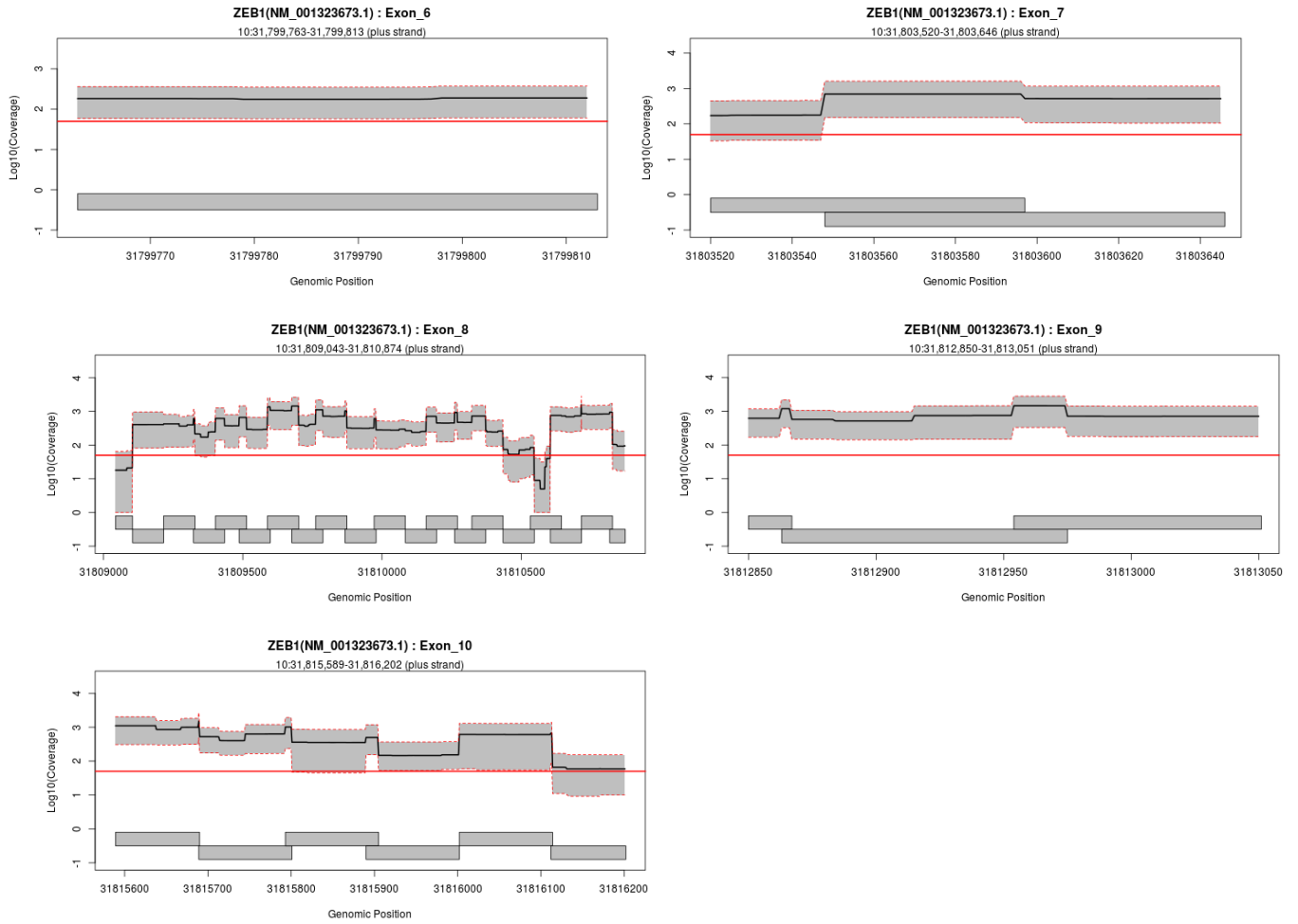




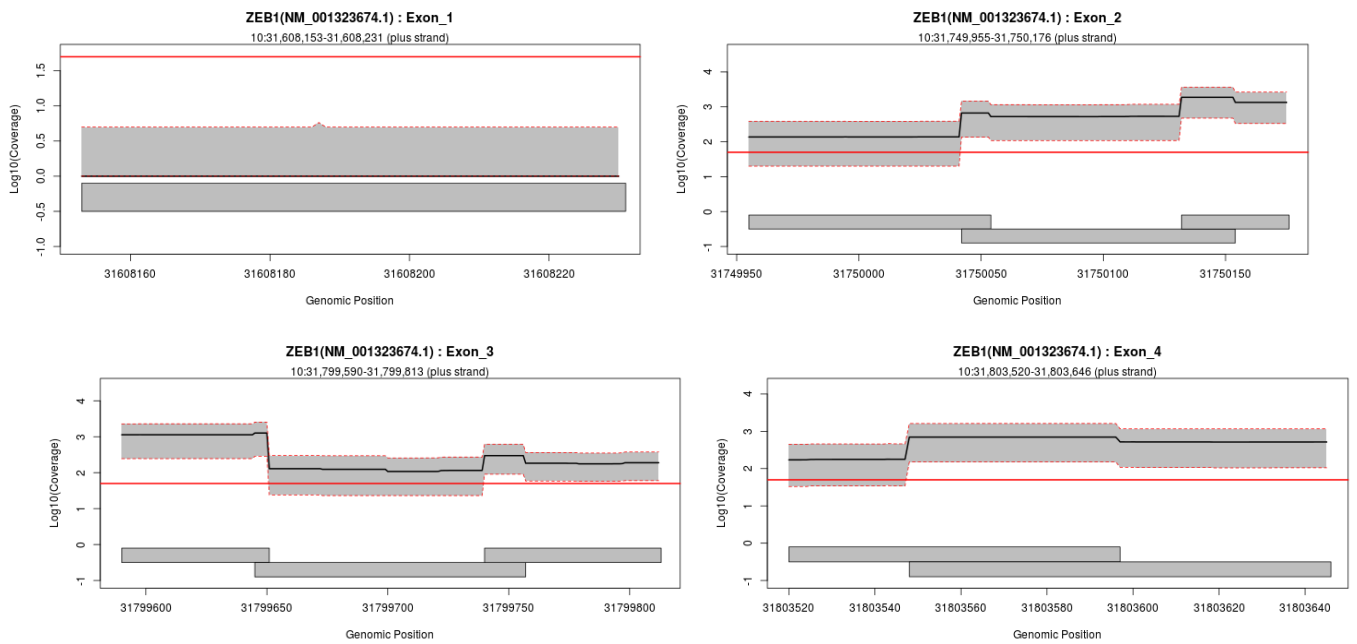
### Exon Overview: ZEB1 : NM\_001323672.1

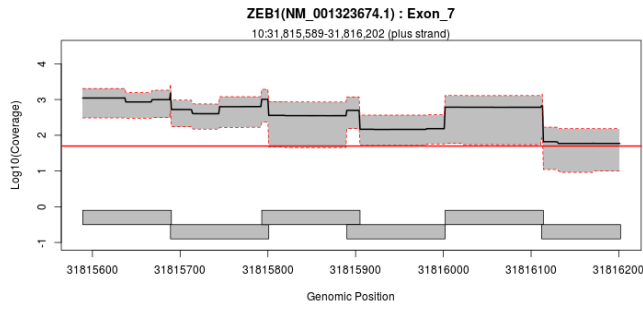
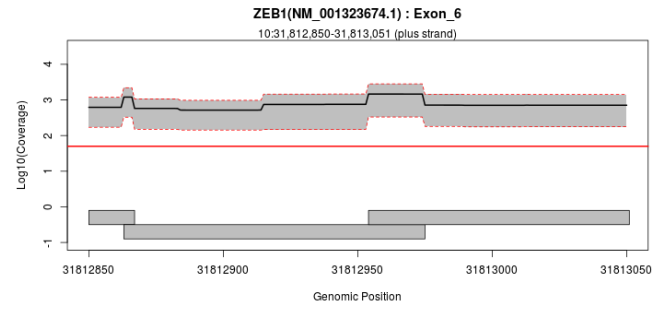
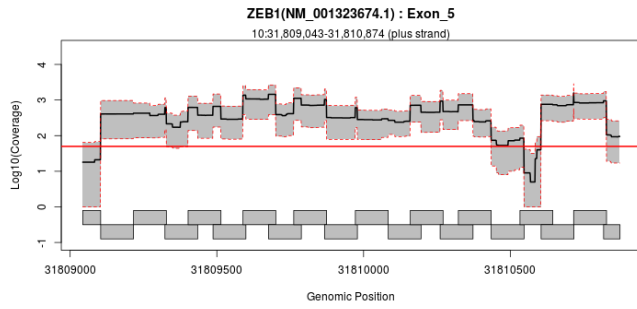


## Exon Overview: ZEB1 : NM\_001323673.1

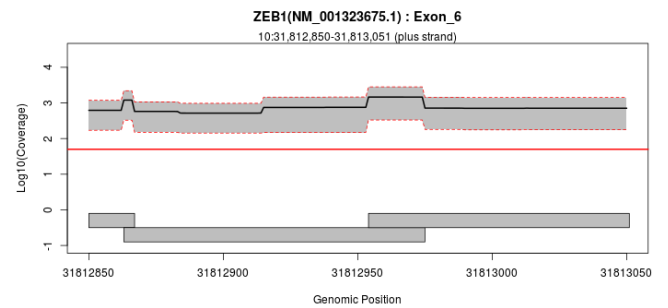
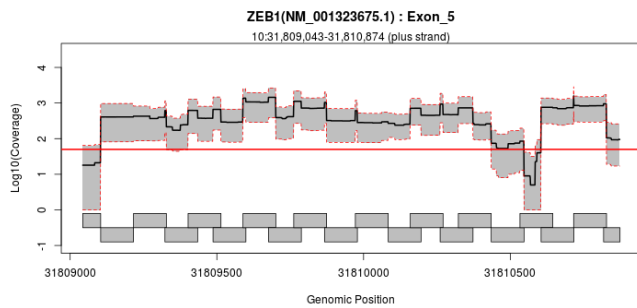
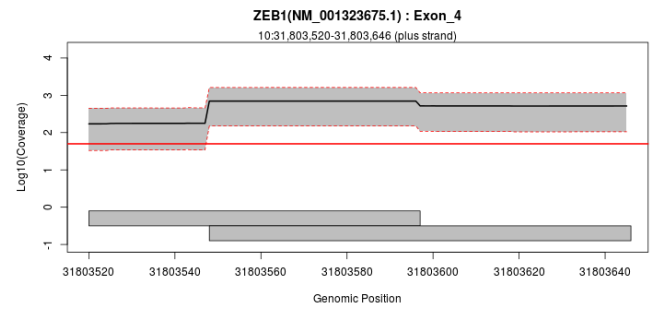
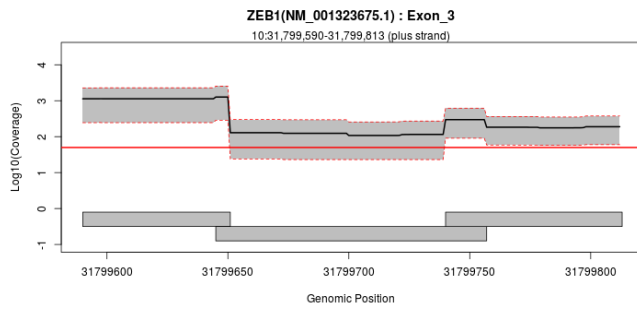
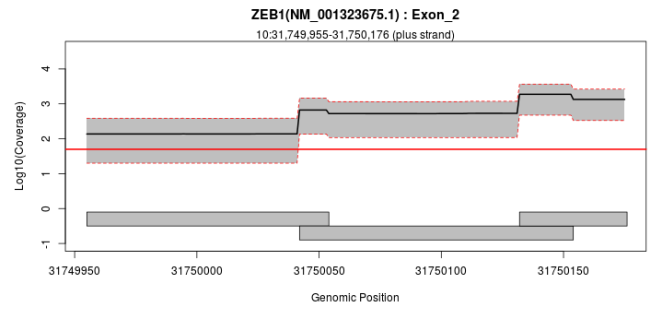
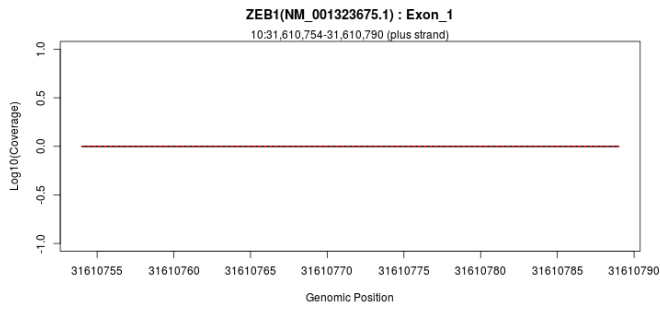


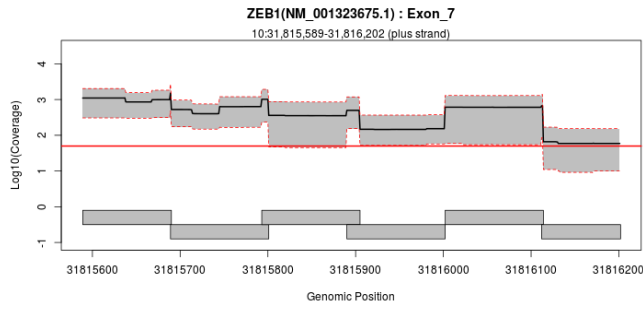
## Exon Overview: ZEB1 : NM\_001323674.1



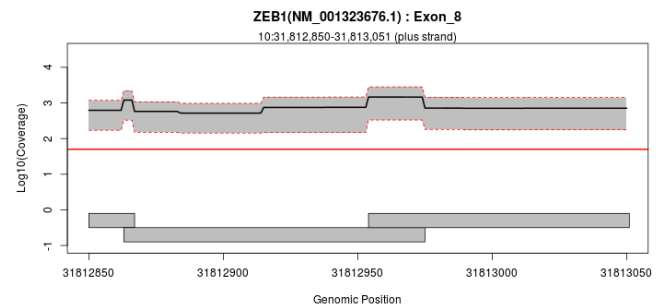
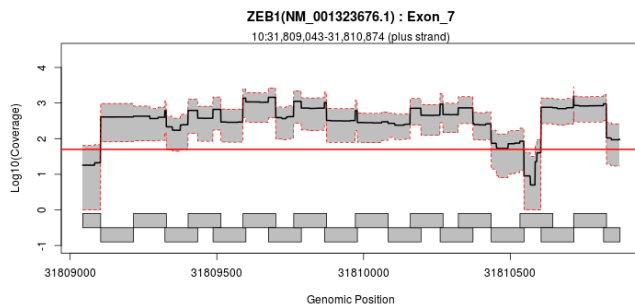
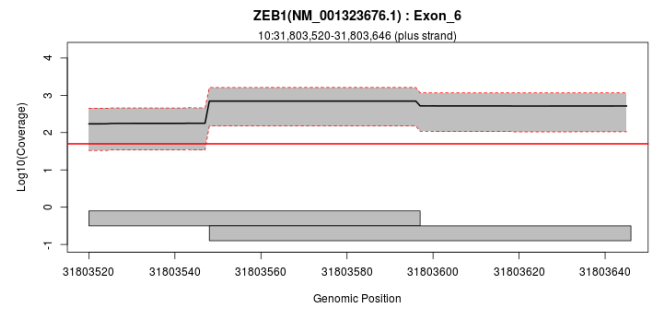
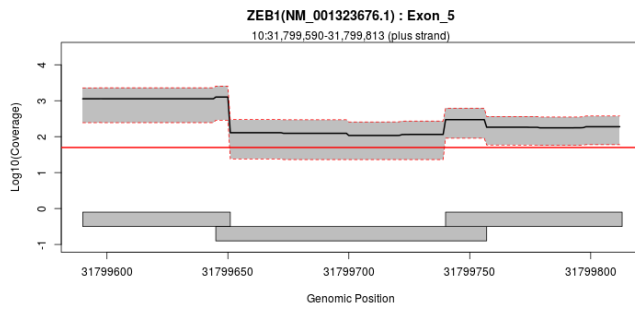
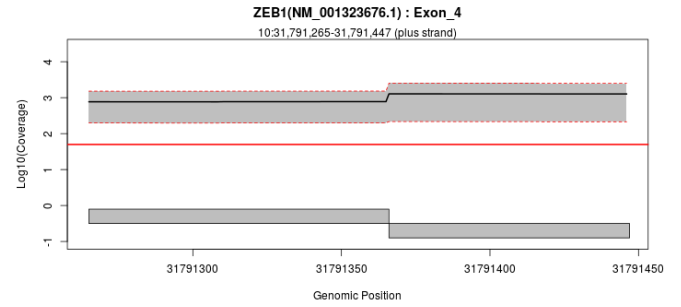
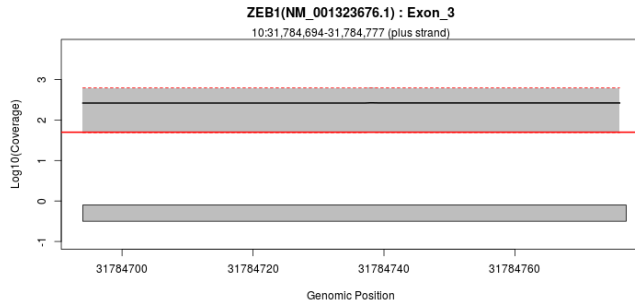
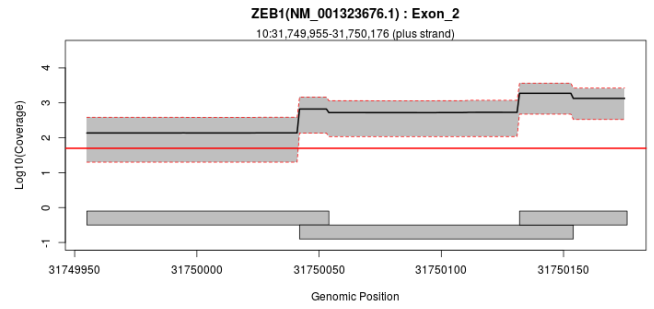
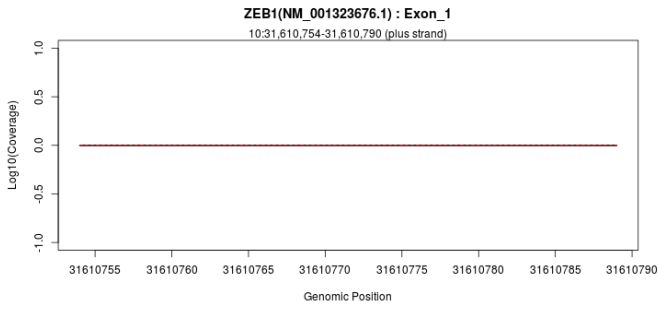


## Exon Overview: ZEB1 : NM\_001323675.1

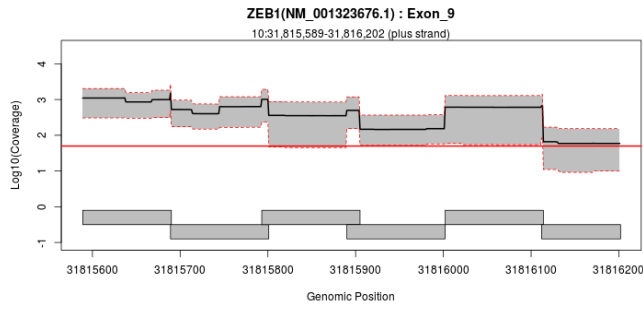




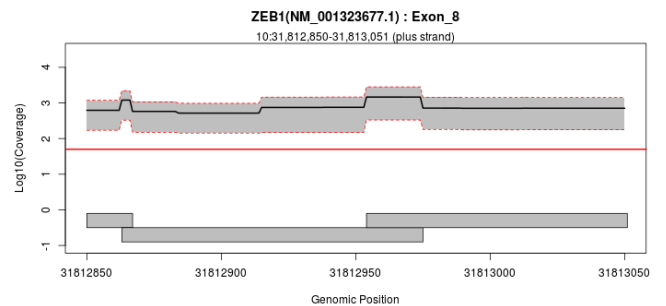
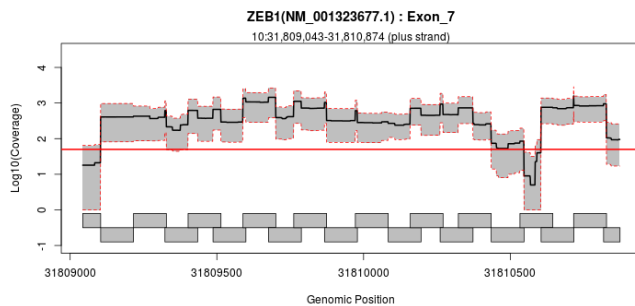
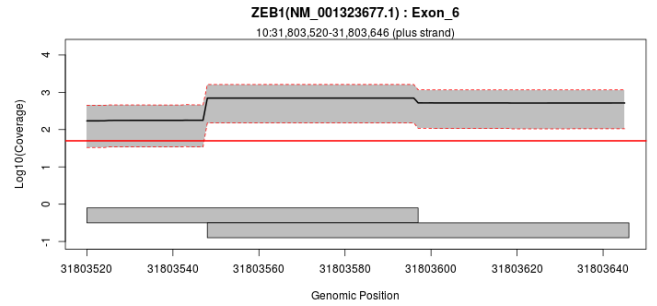
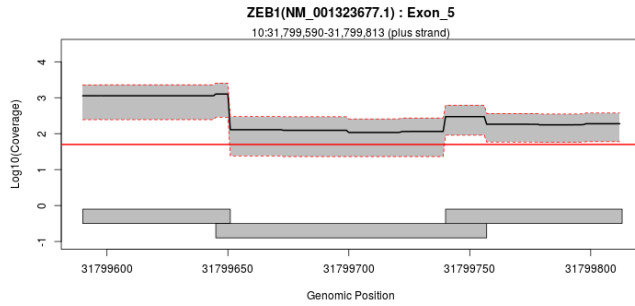
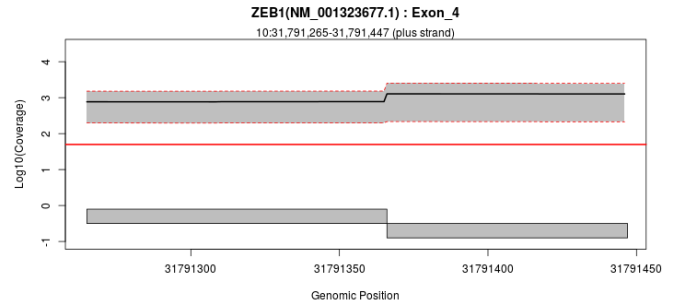
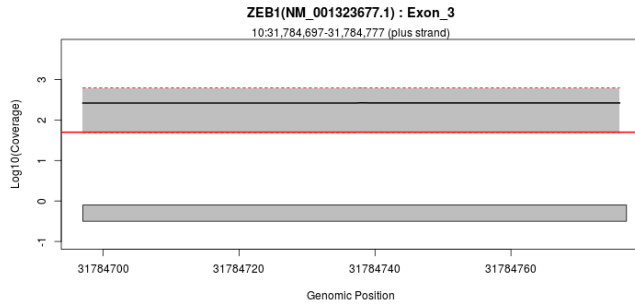
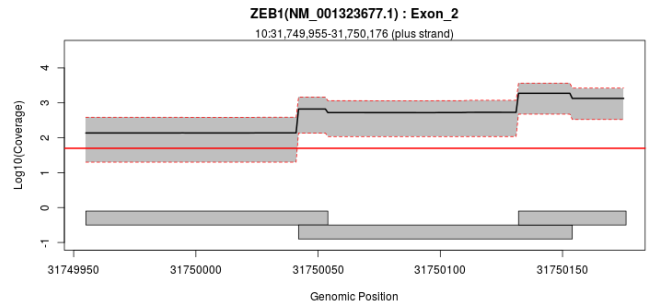
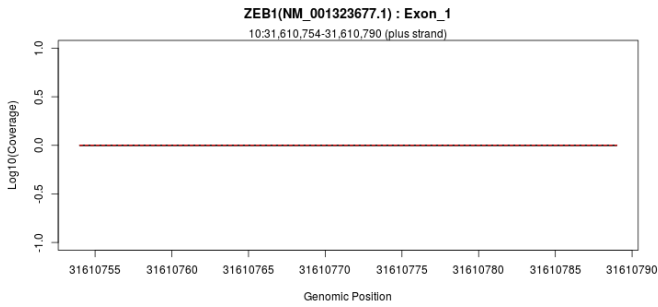
**Exon Overview: ZEB1 : NM\_001323676.1**

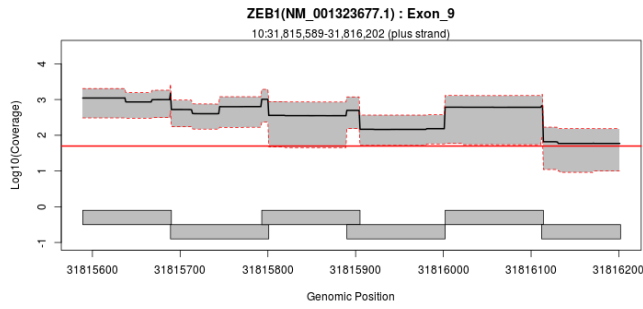




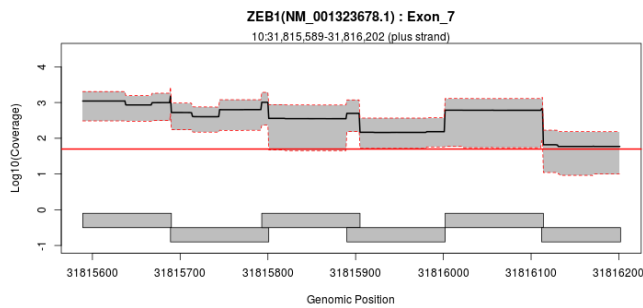
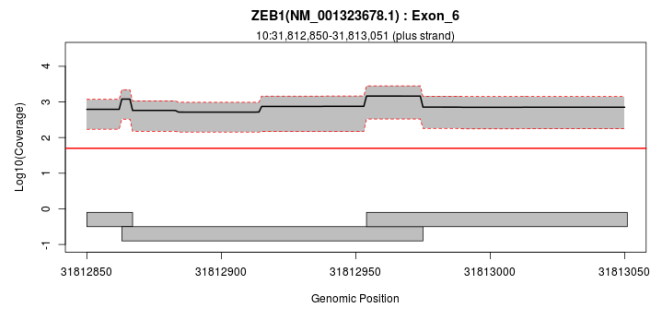
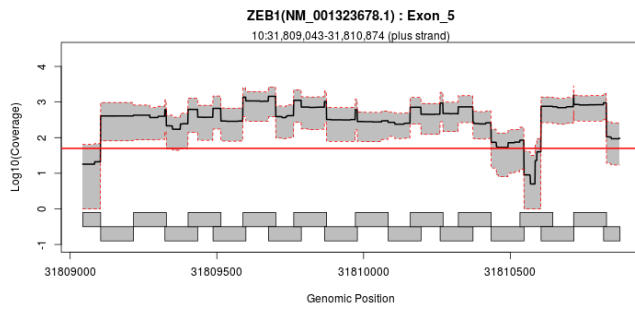
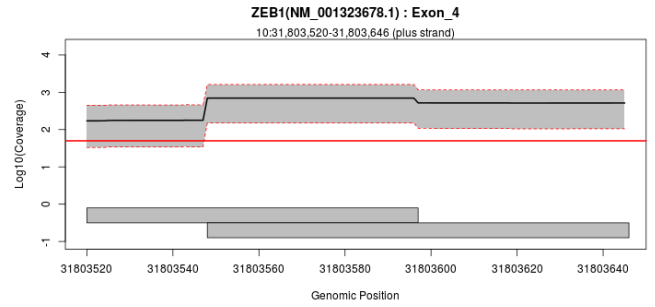
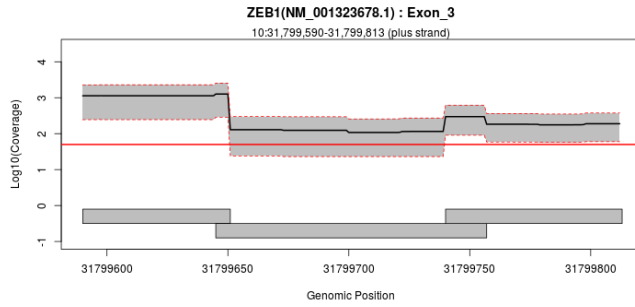
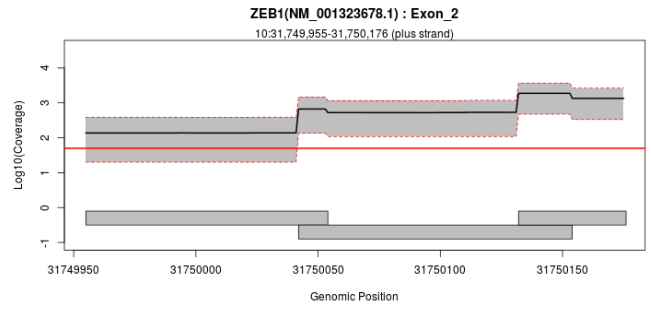
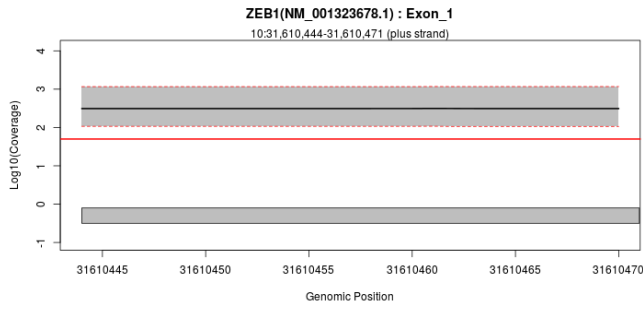


**Exon Overview: ZEB1 : NM\_001323677.1**

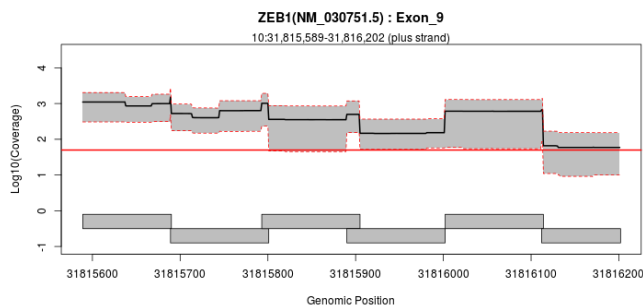
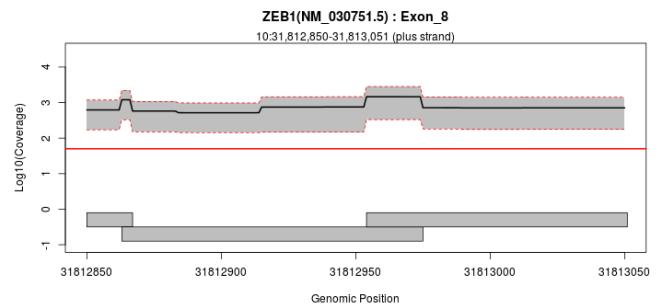
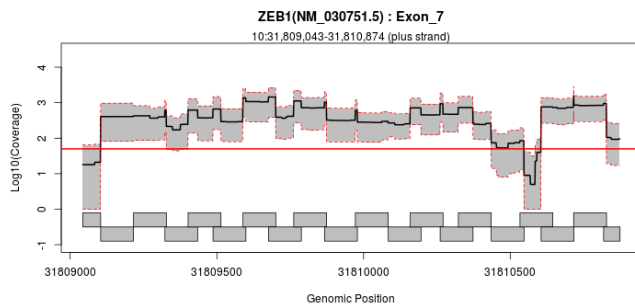
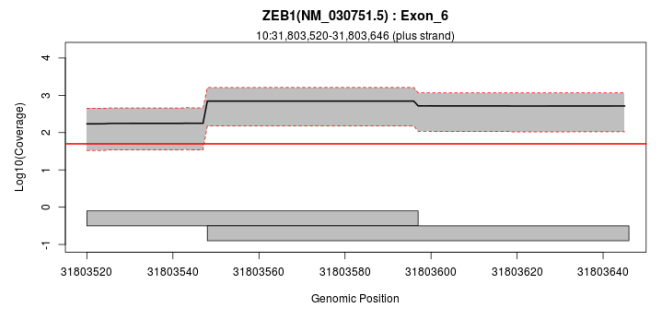
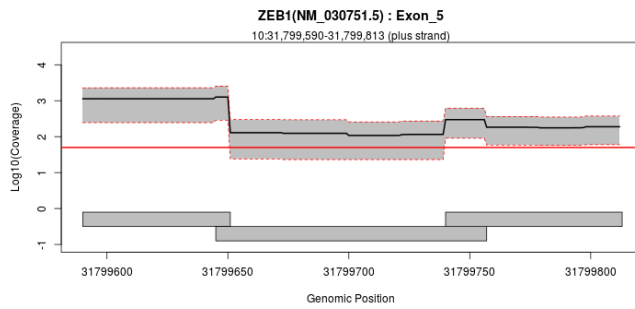
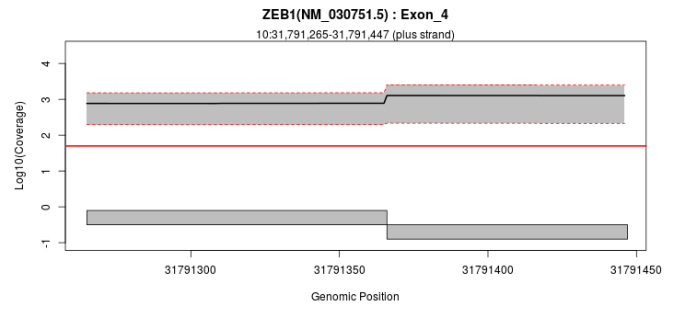
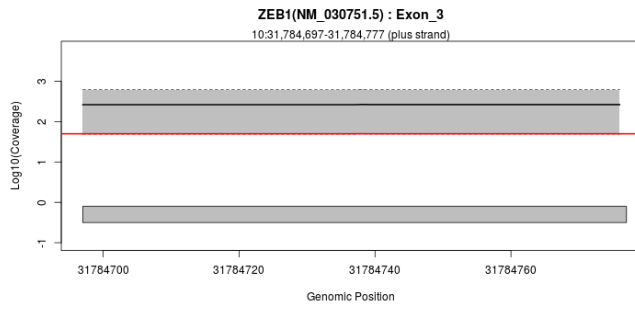
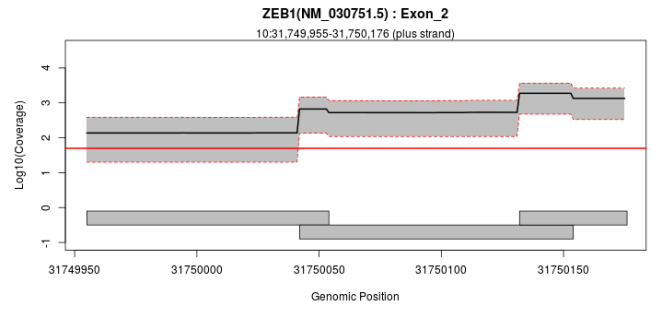
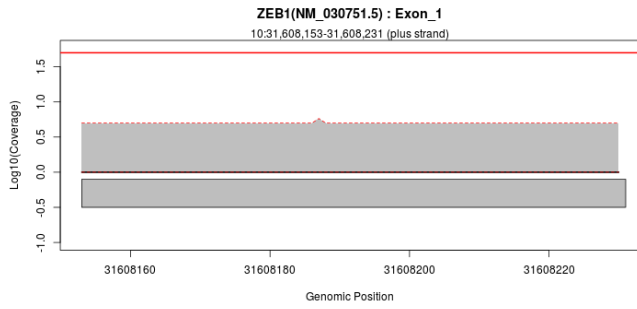




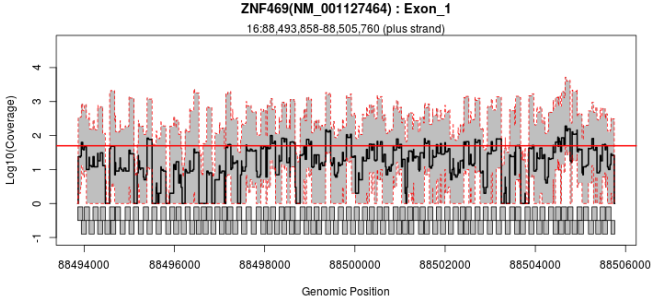
**Exon Overview: ZEB1 : NM\_001323678.1**



# Exon Overview: ZEB1 : NM\_030751.5



# Exon Overview: ZNF469 : NM\_001127464



## Regions Failing minimal coverage criteria

Criteria are defined as a minimal coverage of 50X in at least 20% of samples (equals to 313).

Gene(Transcript) Exon	Region	Size (bp)	Avg.Coverage	Avg.%Samples.Failing
COL12A1(NM_004370.5) Exon_12	6:75,887,478-75,887,529	52	1.53	25.68
COL12A1(NM_004370.5) Exon_16	6:75,865,510-75,865,580	71	5.54	83.18
COL12A1(NM_004370.5) Exon_2	6:75,912,516-75,912,516	1	3.86	20.66
COL12A1(NM_004370.5) Exon_27	6:75,851,884-75,851,887	4	0	100
COL12A1(NM_004370.5) Exon_3	6:75,904,600-75,904,639	40	13.79	82.89
COL12A1(NM_004370.5) Exon_30	6:75,848,026-75,848,066	41	4.87	92.62
COL12A1(NM_004370.5) Exon_46	6:75,828,749-75,828,769	21	3.23	94.04
COL12A1(NM_004370.5) Exon_50	6:75,823,308-75,823,391	84	0.98	34.56
COL12A1(NM_004370.5) Exon_51	6:75,822,910-75,822,996	87	5.58	49.54
COL12A1(NM_004370.5) Exon_52	6:75,818,856-75,818,893	38	16.6	83.62
COL12A1(NM_004370.5) Exon_55	6:75,813,463-75,813,536	74	1.45	23.4
COL12A1(NM_004370.5) Exon_58	6:75,811,306-75,811,379	74	1.81	21.24
COL12A1(NM_004370.5) Exon_66	6:75,796,253-75,796,283	31	8.88	84.61
COL12A1(NM_080645.2) Exon_12	6:75,851,884-75,851,887	4	0	100
COL12A1(NM_080645.2) Exon_15	6:75,848,026-75,848,066	41	4.87	92.62
COL12A1(NM_080645.2) Exon_2	6:75,912,516-75,912,516	1	3.86	20.66
COL12A1(NM_080645.2) Exon_31	6:75,828,749-75,828,769	21	3.23	94.04
COL12A1(NM_080645.2) Exon_35	6:75,823,308-75,823,391	84	0.98	34.56
COL12A1(NM_080645.2) Exon_36	6:75,822,910-75,822,996	87	5.58	49.54
COL12A1(NM_080645.2) Exon_37	6:75,818,856-75,818,893	38	16.6	83.62
COL12A1(NM_080645.2) Exon_40	6:75,813,463-75,813,536	74	1.45	23.4
COL12A1(NM_080645.2) Exon_43	6:75,811,306-75,811,379	74	1.81	21.24
COL12A1(NM_080645.2) Exon_51	6:75,796,253-75,796,283	31	8.88	84.61
COL2A1(NM_001844.4) Exon_14	12:48,387,582-48,387,588	7	5.57	91.65
COL2A1(NM_001844.4) Exon_37	12:48,375,116-48,375,189	74	1.17	32.44
COL2A1(NM_001844.4) Exon_41	12:48,373,357-48,373,357	1	0.57	42.35
COL2A1(NM_001844.4) Exon_42	12:48,372,496-48,372,533	38	1.64	22.32
COL2A1(NM_001844.4) Exon_48	12:48,370,588-48,370,624	37	3.58	20.45
COL2A1(NM_001844.4) Exon_52	12:48,368,631-48,368,655	25	8.54	82.16
COL2A1(NM_001844.4) Exon_7	12:48,391,387-48,391,418	32	4.4	88.72
COL2A1(NM_033150.2) Exon_13	12:48,387,582-48,387,588	7	5.57	91.65
COL2A1(NM_033150.2) Exon_36	12:48,375,116-48,375,189	74	1.17	32.44
COL2A1(NM_033150.2) Exon_40	12:48,373,357-48,373,357	1	0.57	42.35
COL2A1(NM_033150.2) Exon_41	12:48,372,496-48,372,533	38	1.64	22.32
COL2A1(NM_033150.2) Exon_47	12:48,370,588-48,370,624	37	3.58	20.45
COL2A1(NM_033150.2) Exon_51	12:48,368,631-48,368,655	25	8.54	82.16
COL2A1(NM_033150.2) Exon_6	12:48,391,387-48,391,418	32	4.4	88.72
COL5A1(NM_000093.4) Exon_1	9:137,534,024-137,534,152	129	0.48	90.12
COL5A1(NM_000093.4) Exon_23	9:137,658,836-137,658,836	1	1.89	100
COL5A1(NM_000093.4) Exon_43	9:137,701,125-137,701,146	22	7.29	94.5
COL5A1(NM_000093.4) Exon_62	9:137,716,664-137,716,711	48	1.63	21.88
COL5A1(NM_001278074.1) Exon_1	9:137,534,024-137,534,152	129	0.48	90.12
COL5A1(NM_001278074.1) Exon_23	9:137,658,836-137,658,836	1	1.89	100
COL5A1(NM_001278074.1) Exon_43	9:137,701,125-137,701,146	22	7.29	94.5
COL5A1(NM_001278074.1) Exon_62	9:137,716,664-137,716,711	48	1.63	21.88
COL5A3(NM_015719.3) Exon_1	19:10,121,045-10,121,071	27	1.69	22.82
COL5A3(NM_015719.3) Exon_11	19:10,108,037-10,108,089	53	3.63	89.84
COL5A3(NM_015719.3) Exon_17	19:10,104,419-10,104,419	1	0.47	100
COL5A3(NM_015719.3) Exon_18	19:10,104,358-10,104,358	1	3.06	89.48
COL5A3(NM_015719.3) Exon_3	19:10,116,462-10,116,499	38	4.66	100
COL5A3(NM_015719.3) Exon_30	19:10,097,058-10,097,098	41	14.35	77.62
COL5A3(NM_015719.3) Exon_36	19:10,090,660-10,090,733	74	10.9	81.37
COL5A3(NM_015719.3) Exon_41	19:10,089,240-10,089,241	2	15.16	81.09
COL5A3(NM_015719.3) Exon_51	19:10,083,585-10,083,589	5	13.06	41.73
COL5A3(NM_015719.3) Exon_67	19:10,071,116-10,071,149	34	5.94	100
COL5A3(NM_015719.3) Exon_67	19:10,071,153-10,071,182	30	3.47	20.61
DOCK1(NM_001290223.1) Exon_12	10:128,810,486-128,810,548	63	0	100
DOCK1(NM_001290223.1) Exon_14	10:128,821,454-128,821,529	76	8.07	85.61
DOCK1(NM_001290223.1) Exon_17	10:128,829,904-128,829,958	55	3.77	93.57
DOCK1(NM_001290223.1) Exon_21	10:128,841,429-128,841,438	10	3.05	90.07
DOCK1(NM_001290223.1) Exon_36	10:129,178,336-129,178,393	58	12.45	83.01
DOCK1(NM_001290223.1) Exon_36	10:129,178,395-129,178,396	2	2.89	100
DOCK1(NM_001380.4) Exon_14	10:128,821,454-128,821,529	76	8.07	85.61
DOCK1(NM_001380.4) Exon_17	10:128,829,904-128,829,958	55	3.77	93.57
DOCK1(NM_001380.4) Exon_21	10:128,841,429-128,841,438	10	3.05	90.07
DOCK1(NM_001380.4) Exon_36	10:129,178,336-129,178,393	58	12.45	83.01
DOCK1(NM_001380.4) Exon_36	10:129,178,395-129,178,396	2	2.89	100
DOCK9(NM_001130048.1) Exon_19	13:99,538,776-99,538,824	49	11.41	83.69
DOCK9(NM_001130048.1) Exon_28	13:99,520,570-99,520,590	21	12.7	84.23
DOCK9(NM_001130048.1) Exon_30	13:99,519,737-99,519,769	33	13.76	82.53
DOCK9(NM_001130048.1) Exon_31	13:99,515,775-99,515,812	38	0.25	100
DOCK9(NM_001130048.1) Exon_49	13:99,461,367-99,461,392	26	0	100
DOCK9(NM_001130048.1) Exon_50	13:99,460,884-99,460,939	56	16.53	85.57
DOCK9(NM_001130049.1) Exon_1	13:99,738,467-99,738,485	19	1.5	24.96
DOCK9(NM_001130049.1) Exon_1	13:99,738,506-99,738,507	2	1.5	25.16
DOCK9(NM_001130049.1) Exon_19	13:99,538,776-99,538,824	49	11.41	83.69
DOCK9(NM_001130049.1) Exon_28	13:99,520,570-99,520,590	21	12.7	84.23
DOCK9(NM_001130049.1) Exon_30	13:99,519,737-99,519,769	33	13.76	82.53
DOCK9(NM_001130049.1) Exon_31	13:99,515,775-99,515,812	38	0.25	100
DOCK9(NM_001130049.1) Exon_33	13:99,512,584-99,512,584	1	2.46	100
DOCK9(NM_001130050.1) Exon_19	13:99,538,776-99,538,824	49	11.41	83.69
DOCK9(NM_001130050.1) Exon_28	13:99,520,570-99,520,590	21	12.7	84.23
DOCK9(NM_001130050.1) Exon_30	13:99,519,737-99,519,769	33	13.76	82.53
DOCK9(NM_001130050.1) Exon_31	13:99,515,775-99,515,812	38	0.25	100

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Gene(Transcript) Exon	Region	Size (bp)	Avg.Coverage	Avg.%.Samples.Failing
DOCK9(NM_001318849.1) Exon_33	13:99,512,584-99,512,584	1	2.46	100
DOCK9(NM_001318849.1) Exon_1	13:99,738,467-99,738,485	19	1.5	24.96
DOCK9(NM_001318849.1) Exon_1	13:99,738,506-99,738,507	2	1.5	25.16
DOCK9(NM_001318849.1) Exon_19	13:99,538,776-99,538,824	49	11.41	83.69
DOCK9(NM_001318849.1) Exon_28	13:99,520,570-99,520,590	21	12.7	84.23
DOCK9(NM_001318849.1) Exon_30	13:99,519,737-99,519,769	33	13.76	82.53
DOCK9(NM_001318849.1) Exon_31	13:99,515,775-99,515,812	38	0.25	100
DOCK9(NM_001318849.1) Exon_48	13:99,461,367-99,461,392	26	0	100
DOCK9(NM_001318849.1) Exon_49	13:99,460,884-99,460,939	56	16.53	85.57
DOCK9(NM_015296.2) Exon_1	13:99,738,467-99,738,485	19	1.5	24.96
DOCK9(NM_015296.2) Exon_1	13:99,738,506-99,738,507	2	1.5	25.16
DOCK9(NM_015296.2) Exon_19	13:99,538,776-99,538,824	49	11.41	83.69
DOCK9(NM_015296.2) Exon_28	13:99,520,570-99,520,590	21	12.7	84.23
DOCK9(NM_015296.2) Exon_30	13:99,519,737-99,519,769	33	13.76	82.53
DOCK9(NM_015296.2) Exon_31	13:99,515,775-99,515,812	38	0.25	100
DOCK9(NM_015296.2) Exon_49	13:99,461,367-99,461,392	26	0	100
DOCK9(NM_015296.2) Exon_50	13:99,460,884-99,460,939	56	16.53	85.57
EPPK1(NM_031308.3) Exon_2	8:144,940,611-144,940,670	60	0.9	30.65
EPPK1(NM_031308.3) Exon_2	8:144,941,467-144,941,530	64	1.33	23.81
EPPK1(NM_031308.3) Exon_2	8:144,941,663-144,941,741	79	0.82	33.85
EPPK1(NM_031308.3) Exon_2	8:144,942,402-144,942,431	30	1.48	24.82
EPPK1(NM_031308.3) Exon_2	8:144,943,693-144,943,755	63	1.09	30.77
EPPK1(NM_031308.3) Exon_2	8:144,943,892-144,943,982	91	4.86	86.11
EPPK1(NM_031308.3) Exon_2	8:144,944,403-144,944,494	92	0.72	44.73
EPPK1(NM_031308.3) Exon_2	8:144,944,607-144,944,658	52	1.36	22.82
EPPK1(NM_031308.3) Exon_2	8:144,945,272-144,945,284	13	1.29	24.51
EPPK1(NM_031308.3) Exon_2	8:144,945,705-144,945,735	31	0.95	25.5
EPPK1(NM_031308.3) Exon_2	8:144,946,145-144,946,182	38	0.56	52.48
EPPK1(NM_031308.3) Exon_2	8:144,946,567-144,946,647	81	1.22	31.82
FOXO1(NM_002015.3) Exon_1	13:41,239,882-41,240,212	331	0.57	78.16
FOXO1(NM_002015.3) Exon_1	13:41,240,239-41,240,359	121	1.04	38.03
HLA-G(NM_002127.5) Exon_3	6:29,795,935-29,796,002	68	8.29	90.48
HLA-G(NM_002127.5) Exon_4	6:29,796,505-29,796,552	48	7.75	93.28
LAMC3(NM_006059.3) Exon_1	9:133,884,592-133,884,717	126	0.67	36.38
LAMC3(NM_006059.3) Exon_1	9:133,884,910-133,884,984	75	1.1	28.71
LAMC3(NM_006059.3) Exon_11	9:133,928,227-133,928,284	58	1.83	100
LAMC3(NM_006059.3) Exon_21	9:133,951,208-133,951,208	1	18.41	83.55
LAMC3(NM_006059.3) Exon_3	9:133,907,431-133,907,467	37	1.62	21.82
LOXHD1(NM_001145472.2) Exon_16	18:44,098,082-44,098,105	24	10.06	29.63
LOXHD1(NM_001145472.2) Exon_16	18:44,098,107-44,098,127	21	10.66	20.18
LOXHD1(NM_001145472.2) Exon_19	18:44,085,862-44,085,863	2	5.98	20.7
LOXHD1(NM_001145472.2) Exon_21	18:44,064,972-44,064,987	16	6.88	72.94
LOXHD1(NM_001145472.2) Exon_21	18:44,065,064-44,065,124	61	4.91	24.11
LOXHD1(NM_001145473.2) Exon_2	18:44,098,082-44,098,105	24	10.06	29.63
LOXHD1(NM_001145473.2) Exon_2	18:44,098,107-44,098,127	21	10.66	20.18
LOXHD1(NM_001145473.2) Exon_5	18:44,085,862-44,085,863	2	5.98	20.7
LOXHD1(NM_001145473.2) Exon_7	18:44,064,972-44,064,987	16	6.88	72.94
LOXHD1(NM_001145473.2) Exon_7	18:44,065,064-44,065,124	61	4.91	24.11
LOXHD1(NM_001173129.1) Exon_2	18:44,098,082-44,098,105	24	10.06	29.63
LOXHD1(NM_001173129.1) Exon_2	18:44,098,107-44,098,127	21	10.66	20.18
LOXHD1(NM_001173129.1) Exon_5	18:44,085,862-44,085,863	2	5.98	20.7
LOXHD1(NM_001173129.1) Exon_7	18:44,064,972-44,064,987	16	6.88	72.94
LOXHD1(NM_001173129.1) Exon_7	18:44,065,064-44,065,124	61	4.91	24.11
LOXHD1(NM_001173129.1) Exon_7	18:44,098,082-44,098,105	24	10.06	29.63
LOXHD1(NM_001173129.1) Exon_7	18:44,098,107-44,098,127	21	10.66	20.18
LOXHD1(NM_001308013.1) Exon_14	18:44,098,082-44,098,105	24	10.06	29.63
LOXHD1(NM_001308013.1) Exon_14	18:44,098,107-44,098,127	21	10.66	20.18
LOXHD1(NM_001308013.1) Exon_17	18:44,085,862-44,085,863	2	5.98	20.7
LOXHD1(NM_001308013.1) Exon_19	18:44,064,972-44,064,987	16	6.88	72.94
LOXHD1(NM_001308013.1) Exon_19	18:44,065,064-44,065,124	61	4.91	24.11
LOXHD1(NM_144612.6) Exon_19	18:44,140,036-44,140,087	52	3.37	95.5
LOXHD1(NM_144612.6) Exon_33	18:44,098,082-44,098,105	24	10.06	29.63
LOXHD1(NM_144612.6) Exon_33	18:44,098,107-44,098,127	21	10.66	20.18
LOXHD1(NM_144612.6) Exon_36	18:44,085,862-44,085,863	2	5.98	20.7
LOXHD1(NM_144612.6) Exon_38	18:44,064,972-44,064,987	16	6.88	72.94
LOXHD1(NM_144612.6) Exon_38	18:44,065,064-44,065,124	61	4.91	24.11
MIR184.nc(NR_029705.1) Exon_1	15:79,502,120-79,502,120	1	0	100
MMP9(NM_004994.2) Exon_2	20:44,638,744-44,638,747	4	2.59	89.48
MMP9(NM_004994.2) Exon_5	20:44,639,931-44,639,931	1	1.81	19.96
MMP9(NM_004994.2) Exon_5	20:44,639,933-44,639,961	29	1.79	21.16
MMP9(NM_004994.2) Exon_7	20:44,640,805-44,640,861	57	1.82	22.61
MMP9(NM_004994.2) Exon_9	20:44,641,884-44,641,967	84	1.19	26.78
MMP9(NM_004994.2) Exon_9	20:44,641,992-44,642,092	101	1.04	100
NLRP1(NM_001033053.2) Exon_16	17:5,405,124-5,405,124	1	2.06	100
NLRP1(NM_001033053.2) Exon_4	17:5,462,149-5,462,163	15	15.34	81.86
NLRP1(NM_001033053.2) Exon_4	17:5,462,165-5,462,201	37	19.56	86.52
NLRP1(NM_014922.4) Exon_4	17:5,462,149-5,462,163	15	15.34	81.86
NLRP1(NM_014922.4) Exon_4	17:5,462,165-5,462,201	37	19.56	86.52
NLRP1(NM_033004.3) Exon_4	17:5,462,149-5,462,163	15	15.34	81.86
NLRP1(NM_033004.3) Exon_4	17:5,462,165-5,462,201	37	19.56	86.52
NLRP1(NM_033006.3) Exon_4	17:5,462,149-5,462,163	15	15.34	81.86
NLRP1(NM_033006.3) Exon_4	17:5,462,165-5,462,201	37	19.56	86.52
NLRP1(NM_033007.3) Exon_4	17:5,462,149-5,462,163	15	15.34	81.86
NLRP1(NM_033007.3) Exon_4	17:5,462,165-5,462,201	37	19.56	86.52
P3H4(NM_006455.2) Exon_1	17:39,967,813-39,967,913	101	0.66	41.92
P3H4(NM_006455.2) Exon_1	17:39,967,917-39,968,023	107	1.14	28.67
P3H4(NM_006455.2) Exon_1	17:39,968,026-39,968,076	51	1.32	23.07
P3H4(NM_006455.2) Exon_3	17:39,967,195-39,967,208	14	1.44	24.46
P3H4(NM_006455.2) Exon_6	17:39,963,141-39,963,141	1	1.52	22.9
PARP1(NM_001618.3) Exon_11	1:226,567,352-226,567,352	1	0.25	100
PARP2(NM_001042618.1) Exon_15	14:20,825,545-20,825,622	78	1.51	27.12

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Gene(Transcript) Exon	Region	Size (bp)	Avg.Coverage	Avg.%Samples.Failing
PARP2(NM_005484.3) Exon_15	14:20,825,545-20,825,622	78	1.51	27.12
RDH13(NM_001145971.1) Exon_1	19:55,574,400-55,574,406	7	0.16	67.78
SLC4A11(NM_001174089.1) Exon_10	20:3,211,569-3,211,611	43	0.63	100
SLC4A11(NM_001174089.1) Exon_14	20:3,210,211-3,210,293	83	0.97	33.81
SLC4A11(NM_001174089.1) Exon_16	20:3,209,731-3,209,802	72	0.74	100
SLC4A11(NM_001174090.1) Exon_1	20:3,218,529-3,218,583	55	0.9	25.68
SLC4A11(NM_001174090.1) Exon_10	20:3,211,569-3,211,611	43	0.63	100
SLC4A11(NM_001174090.1) Exon_14	20:3,210,211-3,210,293	83	0.97	33.81
SLC4A11(NM_001174090.1) Exon_16	20:3,209,731-3,209,802	72	0.74	100
SLC4A11(NM_032034.3) Exon_13	20:3,210,211-3,210,293	83	0.97	33.81
SLC4A11(NM_032034.3) Exon_15	20:3,209,731-3,209,802	72	0.74	100
SLC4A11(NM_032034.3) Exon_9	20:3,211,569-3,211,611	43	0.63	100
SOD1(NM_000454.4) Exon_1	21:33,032,130-33,032,164	35	1.29	24
TF(NM_001063.3) Exon_13	3:133,486,863-133,486,905	43	2.61	75.78
TGFBI(NM_000358.2) Exon_14	5:135,396,513-135,396,536	24	0.83	37.81
TGFBI(NM_000358.2) Exon_4	5:135,382,014-135,382,053	40	10.71	94.74
TNXB(NM_019105.6) Exon_10	6:32,049,135-32,049,154	20	0	100
TNXB(NM_019105.6) Exon_10	6:32,049,441-32,049,460	20	0	100
TNXB(NM_019105.6) Exon_11	6:32,046,800-32,046,819	20	0	100
TNXB(NM_019105.6) Exon_11	6:32,047,133-32,047,152	20	0	100
TNXB(NM_019105.6) Exon_12	6:32,041,414-32,041,486	73	2.14	99.94
TNXB(NM_019105.6) Exon_12	6:32,041,505-32,041,589	85	6.52	49.22
TNXB(NM_019105.6) Exon_12	6:32,041,720-32,041,739	20	0	100
TNXB(NM_019105.6) Exon_13	6:32,039,757-32,039,776	20	0	100
TNXB(NM_019105.6) Exon_13	6:32,039,851-32,039,861	11	0.7	47.58
TNXB(NM_019105.6) Exon_13	6:32,040,066-32,040,085	20	0	100
TNXB(NM_019105.6) Exon_14	6:32,037,894-32,037,913	20	0	100
TNXB(NM_019105.6) Exon_14	6:32,038,144-32,038,177	34	2.13	29.79
TNXB(NM_019105.6) Exon_14	6:32,038,182-32,038,201	20	0	100
TNXB(NM_019105.6) Exon_15	6:32,037,320-32,037,339	20	0	100
TNXB(NM_019105.6) Exon_15	6:32,037,629-32,037,648	20	0	100
TNXB(NM_019105.6) Exon_16	6:32,036,589-32,036,608	20	0	100
TNXB(NM_019105.6) Exon_16	6:32,036,854-32,036,872	19	1.37	20.69
TNXB(NM_019105.6) Exon_16	6:32,036,904-32,036,923	20	0	100
TNXB(NM_019105.6) Exon_17	6:32,036,157-32,036,176	20	0	100
TNXB(NM_019105.6) Exon_17	6:32,036,475-32,036,494	20	0	100
TNXB(NM_019105.6) Exon_18	6:32,035,428-32,035,447	20	0	100
TNXB(NM_019105.6) Exon_18	6:32,035,679-32,035,698	20	0.84	100
TNXB(NM_019105.6) Exon_18	6:32,035,752-32,035,771	20	0	100
TNXB(NM_019105.6) Exon_19	6:32,032,588-32,032,607	20	0	100
TNXB(NM_019105.6) Exon_19	6:32,032,885-32,032,904	20	0	100
TNXB(NM_019105.6) Exon_2	6:32,065,563-32,065,582	20	0	100
TNXB(NM_019105.6) Exon_2	6:32,065,759-32,065,917	159	3.75	74.65
TNXB(NM_019105.6) Exon_2	6:32,065,966-32,065,985	20	0	100
TNXB(NM_019105.6) Exon_20	6:32,029,924-32,029,943	20	0	100
TNXB(NM_019105.6) Exon_20	6:32,030,188-32,030,270	83	3.08	86.97
TNXB(NM_019105.6) Exon_21	6:32,029,164-32,029,183	20	0	100
TNXB(NM_019105.6) Exon_21	6:32,029,353-32,029,358	6	0.15	100
TNXB(NM_019105.6) Exon_21	6:32,029,435-32,029,437	3	3.08	37.95
TNXB(NM_019105.6) Exon_21	6:32,029,488-32,029,507	20	0	100
TNXB(NM_019105.6) Exon_22	6:32,025,825-32,025,844	20	0	100
TNXB(NM_019105.6) Exon_22	6:32,026,158-32,026,177	20	0	100
TNXB(NM_019105.6) Exon_23	6:32,024,353-32,024,372	20	0	100
TNXB(NM_019105.6) Exon_23	6:32,024,671-32,024,690	20	0	100
TNXB(NM_019105.6) Exon_24	6:32,023,618-32,023,637	20	0	100
TNXB(NM_019105.6) Exon_24	6:32,023,942-32,023,961	20	0	100
TNXB(NM_019105.6) Exon_25	6:32,021,155-32,021,174	20	0	100
TNXB(NM_019105.6) Exon_25	6:32,021,473-32,021,492	20	0	100
TNXB(NM_019105.6) Exon_26	6:32,020,437-32,020,456	20	0	100
TNXB(NM_019105.6) Exon_26	6:32,020,761-32,020,780	20	0	100
TNXB(NM_019105.6) Exon_27	6:32,017,765-32,017,784	20	0	100
TNXB(NM_019105.6) Exon_27	6:32,018,089-32,018,108	20	0	100
TNXB(NM_019105.6) Exon_28	6:32,017,037-32,017,056	20	0	100
TNXB(NM_019105.6) Exon_28	6:32,017,355-32,017,374	20	0	100
TNXB(NM_019105.6) Exon_29	6:32,016,130-32,016,149	20	0	100
TNXB(NM_019105.6) Exon_29	6:32,016,418-32,016,437	20	0	100
TNXB(NM_019105.6) Exon_3	6:32,063,378-32,063,397	20	0	100
TNXB(NM_019105.6) Exon_3	6:32,063,468-32,063,499	32	1.56	23.28
TNXB(NM_019105.6) Exon_3	6:32,063,636-32,063,640	5	2.06	20
TNXB(NM_019105.6) Exon_3	6:32,063,643-32,063,683	41	1.72	24.33
TNXB(NM_019105.6) Exon_3	6:32,064,020-32,064,047	28	12.08	78
TNXB(NM_019105.6) Exon_3	6:32,065,217-32,065,236	20	0	100
TNXB(NM_019105.6) Exon_30	6:32,015,501-32,015,520	20	0	100
TNXB(NM_019105.6) Exon_30	6:32,015,780-32,015,799	20	0	100
TNXB(NM_019105.6) Exon_31	6:32,013,942-32,013,961	20	0	100
TNXB(NM_019105.6) Exon_31	6:32,014,224-32,014,243	20	0	100
TNXB(NM_019105.6) Exon_32	6:32,012,773-32,012,803	31	0.49	100
TNXB(NM_019105.6) Exon_32	6:32,012,990-32,013,019	30	1.23	97.22
TNXB(NM_019105.6) Exon_32	6:32,013,080-32,013,113	34	0.67	100
TNXB(NM_019105.6) Exon_33	6:32,012,148-32,012,167	20	0	100
TNXB(NM_019105.6) Exon_33	6:32,012,212-32,012,262	51	0.13	100
TNXB(NM_019105.6) Exon_33	6:32,012,323-32,012,373	51	0.12	100
TNXB(NM_019105.6) Exon_33	6:32,012,433-32,012,473	41	0.15	100
TNXB(NM_019105.6) Exon_33	6:32,012,484-32,012,503	20	0	100
TNXB(NM_019105.6) Exon_34	6:32,011,774-32,011,793	20	0	100
TNXB(NM_019105.6) Exon_34	6:32,011,814-32,011,816	3	0.36	100
TNXB(NM_019105.6) Exon_34	6:32,011,875-32,011,916	42	0.15	82.41
TNXB(NM_019105.6) Exon_35	6:32,011,516-32,011,535	20	0	100
TNXB(NM_019105.6) Exon_35	6:32,011,613-32,011,633	21	0.55	44.21

Continued on next page

Gene(Transcript) Exon	Region	Size (bp)	Avg.Coverage	Avg.%Samples.Failing
TNXB(NM_019105.6) Exon_35	6:32,011,660-32,011,679	20	0	100
TNXB(NM_019105.6) Exon_36	6:32,011,204-32,011,245	42	0.02	100
TNXB(NM_019105.6) Exon_36	6:32,011,306-32,011,343	38	0.14	100
TNXB(NM_019105.6) Exon_37	6:32,010,942-32,010,961	20	0	100
TNXB(NM_019105.6) Exon_37	6:32,010,968-32,010,984	17	1.29	98.61
TNXB(NM_019105.6) Exon_37	6:32,011,045-32,011,048	4	1.29	94.66
TNXB(NM_019105.6) Exon_37	6:32,011,086-32,011,105	20	0	100
TNXB(NM_019105.6) Exon_38	6:32,010,718-32,010,836	119	0.11	78.31
TNXB(NM_019105.6) Exon_38	6:32,010,849-32,010,868	20	0	100
TNXB(NM_019105.6) Exon_39	6:32,010,466-32,010,485	20	0	100
TNXB(NM_019105.6) Exon_39	6:32,010,510-32,010,553	44	2.04	93.09
TNXB(NM_019105.6) Exon_39	6:32,010,599-32,010,618	20	0	100
TNXB(NM_019105.6) Exon_4	6:32,062,843-32,062,862	20	0	100
TNXB(NM_019105.6) Exon_4	6:32,062,959-32,062,978	20	0	100
TNXB(NM_019105.6) Exon_40	6:32,010,222-32,010,241	20	0	100
TNXB(NM_019105.6) Exon_40	6:32,010,374-32,010,393	20	0	100
TNXB(NM_019105.6) Exon_41	6:32,010,033-32,010,102	70	0.1	99.96
TNXB(NM_019105.6) Exon_41	6:32,010,130-32,010,149	20	0	100
TNXB(NM_019105.6) Exon_42	6:32,009,779-32,009,798	20	0	100
TNXB(NM_019105.6) Exon_42	6:32,009,846-32,009,888	43	0.32	99.02
TNXB(NM_019105.6) Exon_42	6:32,009,941-32,009,960	20	0	100
TNXB(NM_019105.6) Exon_43	6:32,009,538-32,009,557	20	0	100
TNXB(NM_019105.6) Exon_43	6:32,009,588-32,009,625	38	0.12	100
TNXB(NM_019105.6) Exon_43	6:32,009,686-32,009,721	36	0.21	100
TNXB(NM_019105.6) Exon_44	6:32,009,116-32,009,166	51	0.04	100
TNXB(NM_019105.6) Exon_44	6:32,009,218-32,009,237	20	0	100
TNXB(NM_019105.6) Exon_5	6:32,056,990-32,057,009	20	0	100
TNXB(NM_019105.6) Exon_5	6:32,057,147-32,057,166	20	0	100
TNXB(NM_019105.6) Exon_6	6:32,056,552-32,056,571	20	0	100
TNXB(NM_019105.6) Exon_6	6:32,056,816-32,056,835	20	0	100
TNXB(NM_019105.6) Exon_7	6:32,053,517-32,053,536	20	0	100
TNXB(NM_019105.6) Exon_7	6:32,053,649-32,053,692	44	1.78	23.39
TNXB(NM_019105.6) Exon_7	6:32,053,886-32,053,905	20	0	100
TNXB(NM_019105.6) Exon_8	6:32,052,180-32,052,199	20	0	100
TNXB(NM_019105.6) Exon_8	6:32,052,477-32,052,496	20	0	100
TNXB(NM_019105.6) Exon_9	6:32,049,803-32,049,822	20	0	100
TNXB(NM_019105.6) Exon_9	6:32,050,094-32,050,113	20	0	100
TNXB(NM_032470.3) Exon_1	6:32,012,773-32,012,803	31	0.49	100
TNXB(NM_032470.3) Exon_1	6:32,012,990-32,013,006	17	2.19	95.1
TNXB(NM_032470.3) Exon_10	6:32,010,033-32,010,102	70	0.1	99.96
TNXB(NM_032470.3) Exon_10	6:32,010,130-32,010,149	20	0	100
TNXB(NM_032470.3) Exon_11	6:32,009,779-32,009,798	20	0	100
TNXB(NM_032470.3) Exon_11	6:32,009,846-32,009,888	43	0.32	99.02
TNXB(NM_032470.3) Exon_11	6:32,009,941-32,009,960	20	0	100
TNXB(NM_032470.3) Exon_12	6:32,009,538-32,009,557	20	0	100
TNXB(NM_032470.3) Exon_12	6:32,009,588-32,009,625	38	0.12	100
TNXB(NM_032470.3) Exon_12	6:32,009,686-32,009,721	36	0.21	100
TNXB(NM_032470.3) Exon_13	6:32,009,116-32,009,166	51	0.04	100
TNXB(NM_032470.3) Exon_13	6:32,009,218-32,009,237	20	0	100
TNXB(NM_032470.3) Exon_2	6:32,012,148-32,012,167	20	0	100
TNXB(NM_032470.3) Exon_2	6:32,012,212-32,012,262	51	0.13	100
TNXB(NM_032470.3) Exon_2	6:32,012,323-32,012,373	51	0.12	100
TNXB(NM_032470.3) Exon_2	6:32,012,433-32,012,473	41	0.15	100
TNXB(NM_032470.3) Exon_2	6:32,012,484-32,012,503	20	0	100
TNXB(NM_032470.3) Exon_3	6:32,011,774-32,011,793	20	0	100
TNXB(NM_032470.3) Exon_3	6:32,011,814-32,011,816	3	0.36	100
TNXB(NM_032470.3) Exon_3	6:32,011,875-32,011,916	42	0.15	82.41
TNXB(NM_032470.3) Exon_4	6:32,011,516-32,011,535	20	0	100
TNXB(NM_032470.3) Exon_4	6:32,011,613-32,011,633	21	0.55	44.21
TNXB(NM_032470.3) Exon_4	6:32,011,660-32,011,679	20	0	100
TNXB(NM_032470.3) Exon_5	6:32,011,204-32,011,245	42	0.02	100
TNXB(NM_032470.3) Exon_5	6:32,011,306-32,011,343	38	0.14	100
TNXB(NM_032470.3) Exon_6	6:32,010,942-32,010,961	20	0	100
TNXB(NM_032470.3) Exon_6	6:32,010,968-32,010,984	17	1.29	98.61
TNXB(NM_032470.3) Exon_6	6:32,011,045-32,011,048	4	1.29	94.66
TNXB(NM_032470.3) Exon_6	6:32,011,086-32,011,105	20	0	100
TNXB(NM_032470.3) Exon_7	6:32,010,718-32,010,836	119	0.11	78.31
TNXB(NM_032470.3) Exon_7	6:32,010,849-32,010,868	20	0	100
TNXB(NM_032470.3) Exon_8	6:32,010,466-32,010,485	20	0	100
TNXB(NM_032470.3) Exon_8	6:32,010,510-32,010,553	44	2.04	93.09
TNXB(NM_032470.3) Exon_8	6:32,010,599-32,010,618	20	0	100
TNXB(NM_032470.3) Exon_9	6:32,010,222-32,010,241	20	0	100
TNXB(NM_032470.3) Exon_9	6:32,010,374-32,010,393	20	0	100
VSX1(NM_001256271.1) Exon_1	20:25,062,399-25,062,460	62	1.06	36.34
VSX1(NM_001256272.1) Exon_1	20:25,062,399-25,062,460	62	1.06	36.34
VSX1(NM_014588.5) Exon_1	20:25,062,399-25,062,460	62	1.06	36.34
VSX1(NM_199425.2) Exon_1	20:25,062,399-25,062,460	62	1.06	36.34
WNT10A(NM_025216.2) Exon_1	2:219,745,787-219,745,840	54	0.77	35.06
WNT10A(NM_025216.2) Exon_4	2:219,757,570-219,757,632	63	0.56	41.97
WNT10A(NM_025216.2) Exon_4	2:219,757,653-219,757,755	103	1.69	93.73
WNT10A(NM_025216.2) Exon_4	2:219,757,786-219,757,849	64	1.14	26.45
WNT10A(NM_025216.2) Exon_4	2:219,757,888-219,757,952	65	1.22	25.92
ZEB1(NM_001128128.2) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001174093.1) Exon_1	10:31,608,154-31,608,231	78	0.43	100
ZEB1(NM_001174093.1) Exon_6	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001174094.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001174095.1) Exon_1	10:31,608,154-31,608,231	78	0.43	100
ZEB1(NM_001174095.1) Exon_6	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001174096.1) Exon_1	10:31,608,154-31,608,231	78	0.43	100

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Gene(Transcript) Exon	Region	Size (bp)	Avg.Coverage	Avg.%Samples.Failing
ZEB1(NM_001174096.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323638.1) Exon_11	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323641.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323642.1) Exon_9	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323643.1) Exon_9	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323644.1) Exon_6	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323645.1) Exon_8	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323646.1) Exon_10	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323647.1) Exon_8	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323648.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323649.1) Exon_8	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323650.1) Exon_10	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323651.1) Exon_8	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323652.1) Exon_9	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323653.1) Exon_9	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323654.1) Exon_9	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323655.1) Exon_9	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323656.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323657.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323658.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323659.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323660.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323661.1) Exon_8	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323662.1) Exon_9	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323663.1) Exon_10	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323664.1) Exon_9	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323665.1) Exon_8	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323666.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323671.1) Exon_8	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323672.1) Exon_9	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323673.1) Exon_8	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323674.1) Exon_1	10:31,608,154-31,608,231	78	0.43	100
ZEB1(NM_001323674.1) Exon_5	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323675.1) Exon_1	10:31,610,755-31,610,790	36	0	100
ZEB1(NM_001323675.1) Exon_5	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323676.1) Exon_1	10:31,610,755-31,610,790	36	0	100
ZEB1(NM_001323676.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323677.1) Exon_1	10:31,610,755-31,610,790	36	0	100
ZEB1(NM_001323677.1) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_001323678.1) Exon_5	10:31,810,548-31,810,585	38	5.65	49.39
ZEB1(NM_030751.5) Exon_1	10:31,608,154-31,608,231	78	0.43	100
ZEB1(NM_030751.5) Exon_7	10:31,810,548-31,810,585	38	5.65	49.39
ZNF469(NM_001127464) Exon_1	16:88,493,859-88,493,868	10	0	100
ZNF469(NM_001127464) Exon_1	16:88,494,051-88,494,110	60	1.61	23.78
ZNF469(NM_001127464) Exon_1	16:88,494,471-88,494,565	95	0.38	99.42
ZNF469(NM_001127464) Exon_1	16:88,494,678-88,494,692	15	0.66	36.44
ZNF469(NM_001127464) Exon_1	16:88,494,748-88,494,748	1	1.73	20.03
ZNF469(NM_001127464) Exon_1	16:88,494,805-88,494,888	84	1.37	23.49
ZNF469(NM_001127464) Exon_1	16:88,494,890-88,494,909	20	2	20.02
ZNF469(NM_001127464) Exon_1	16:88,494,926-88,494,993	68	1.7	22.62
ZNF469(NM_001127464) Exon_1	16:88,495,106-88,495,176	71	1.4	98.17
ZNF469(NM_001127464) Exon_1	16:88,495,313-88,495,394	82	1.43	27.82
ZNF469(NM_001127464) Exon_1	16:88,495,507-88,495,701	195	0.36	54.33
ZNF469(NM_001127464) Exon_1	16:88,495,814-88,495,984	171	0.16	74.12
ZNF469(NM_001127464) Exon_1	16:88,496,097-88,496,238	142	1.43	64.36
ZNF469(NM_001127464) Exon_1	16:88,496,279-88,496,340	62	1.62	21.27
ZNF469(NM_001127464) Exon_1	16:88,496,351-88,496,432	82	1.09	25.63
ZNF469(NM_001127464) Exon_1	16:88,496,545-88,496,718	174	1.22	75.8
ZNF469(NM_001127464) Exon_1	16:88,496,831-88,496,996	166	1.12	58.75
ZNF469(NM_001127464) Exon_1	16:88,497,011-88,497,140	130	0.79	33.37
ZNF469(NM_001127464) Exon_1	16:88,497,253-88,497,267	15	1.68	22.51
ZNF469(NM_001127464) Exon_1	16:88,497,404-88,497,490	87	1.03	47.66
ZNF469(NM_001127464) Exon_1	16:88,497,515-88,497,570	56	1.39	23.98
ZNF469(NM_001127464) Exon_1	16:88,497,834-88,497,916	83	1.25	27.43
ZNF469(NM_001127464) Exon_1	16:88,498,029-88,498,057	29	1.11	33.49
ZNF469(NM_001127464) Exon_1	16:88,498,242-88,498,321	80	0.54	36.71
ZNF469(NM_001127464) Exon_1	16:88,498,564-88,498,565	2	1.28	23.69
ZNF469(NM_001127464) Exon_1	16:88,498,678-88,498,786	109	2.93	84.76
ZNF469(NM_001127464) Exon_1	16:88,499,042-88,499,095	54	1.16	21.61
ZNF469(NM_001127464) Exon_1	16:88,499,210-88,499,240	31	1.67	23.07
ZNF469(NM_001127464) Exon_1	16:88,499,485-88,499,502	18	1.57	22.33
ZNF469(NM_001127464) Exon_1	16:88,499,547-88,499,608	62	1.04	24.13
ZNF469(NM_001127464) Exon_1	16:88,500,015-88,500,167	153	0.63	39.71
ZNF469(NM_001127464) Exon_1	16:88,500,681-88,500,729	49	1.02	29.8
ZNF469(NM_001127464) Exon_1	16:88,500,772-88,500,806	35	1.43	22.78
ZNF469(NM_001127464) Exon_1	16:88,501,113-88,501,164	52	0.49	49.22
ZNF469(NM_001127464) Exon_1	16:88,501,412-88,501,447	36	1.18	24.84
ZNF469(NM_001127464) Exon_1	16:88,502,107-88,502,155	49	0.88	34.19
ZNF469(NM_001127464) Exon_1	16:88,502,200-88,502,287	88	1.11	31.26
ZNF469(NM_001127464) Exon_1	16:88,502,292-88,502,334	43	1.53	24.29
ZNF469(NM_001127464) Exon_1	16:88,502,775-88,502,936	162	0.86	34.45
ZNF469(NM_001127464) Exon_1	16:88,503,248-88,503,431	184	1.17	70.22
ZNF469(NM_001127464) Exon_1	16:88,503,544-88,503,563	20	0.8	25.84
ZNF469(NM_001127464) Exon_1	16:88,503,676-88,503,856	181	1.11	84.45
ZNF469(NM_001127464) Exon_1	16:88,503,969-88,504,033	65	1.39	24.06
ZNF469(NM_001127464) Exon_1	16:88,504,214-88,504,292	79	0.68	40.84
ZNF469(NM_001127464) Exon_1	16:88,505,037-88,505,065	29	1.43	25.99
ZNF469(NM_001127464) Exon_1	16:88,505,751-88,505,760	10	0	100

Total number of failed regions : 439