

Supporting information

Whole-exome sequencing in eccrine porocarcinoma indicates promising therapeutic strategies

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Supporting Information

Supplemental Fig. 1: Immunohistochemistry and Western Blot analyses of BRCA2-expressing cells treated with BRCA2 siRNA confirm BRCA2 antibody specificity.

The BRCA2 expressing prostata carcinoma cells line LNCAP was treated for 48 h with 43 nM BRCA2 siRNA (siBRCA2, 5'-CCAACUUUGUCCUUAACUA-3', Eurofins) or scrambled siRNA (siSCR, 5'-GCAGCUAUAUGAAUGUUGU-3', Eurofins), according to the Lipofectamine RNAiMAX protocols. **a**, In immunohistochemical analyses, LNCAP cells treated with the BRCA2 siRNA show almost no BRCA2 protein expression. Treated cells were resuspended in HistoGel (#HG4000012, ThermoFisher) in accordance with the manufacturers' protocols, then fixed with 4% formalin and embedded in paraffin. One μm sections of these cell blocks were stained for BRCA2 (see Methods section in main text). Representative images at 100x and 400x (inlays) magnification were taken of the siSCR (left panel) and siBRCA2 (right panel) treated cells showing strong and no BRCA2 staining, respectively. **b**, In Western blot analyses, LNCAP cells treated with the BRCA2 siRNA do not display any discernible BRCA2 protein band at 380 kDa. Treated cells were lysed in IP-lysis buffer (50 mM Hepes, pH7.5; 150 mM NaCl; 10% glycerol; 1% Triton-X-100; 1.5 mM MgCl_2 ; 1 mM EDTA). 25 and 50 μg protein were loaded onto 4-20% gradient gels (#456-1083, BioRad) and blotted onto nitrocellulose membranes (#170-4159, BioRad). Membranes were incubated with BRCA2 (#10741) or GAPDH (#2118) antibodies from Cell Signaling at a 1:1000 dilution o/n at 4°C, followed by an incubation with a HRP-conjugated, rabbit secondary antibody (#NA934, GE Healthcare) at 1:3000 dilution for 1 h at RT. Proteins were detected with Dura Extended Duration substrate (#34075, Thermo Fisher Scientific) and visualised using the ImageQuant LAS 4000 detection system (GE Healthcare). Images of the uncut BRCA2- (left panel) and GAPDH- (right panel) stained membranes are shown. *is a non-specific band at 35 kDa detected with the BRCA2 antibody.

Supplemental table 1: Whole-exome sequencing coverage statistics

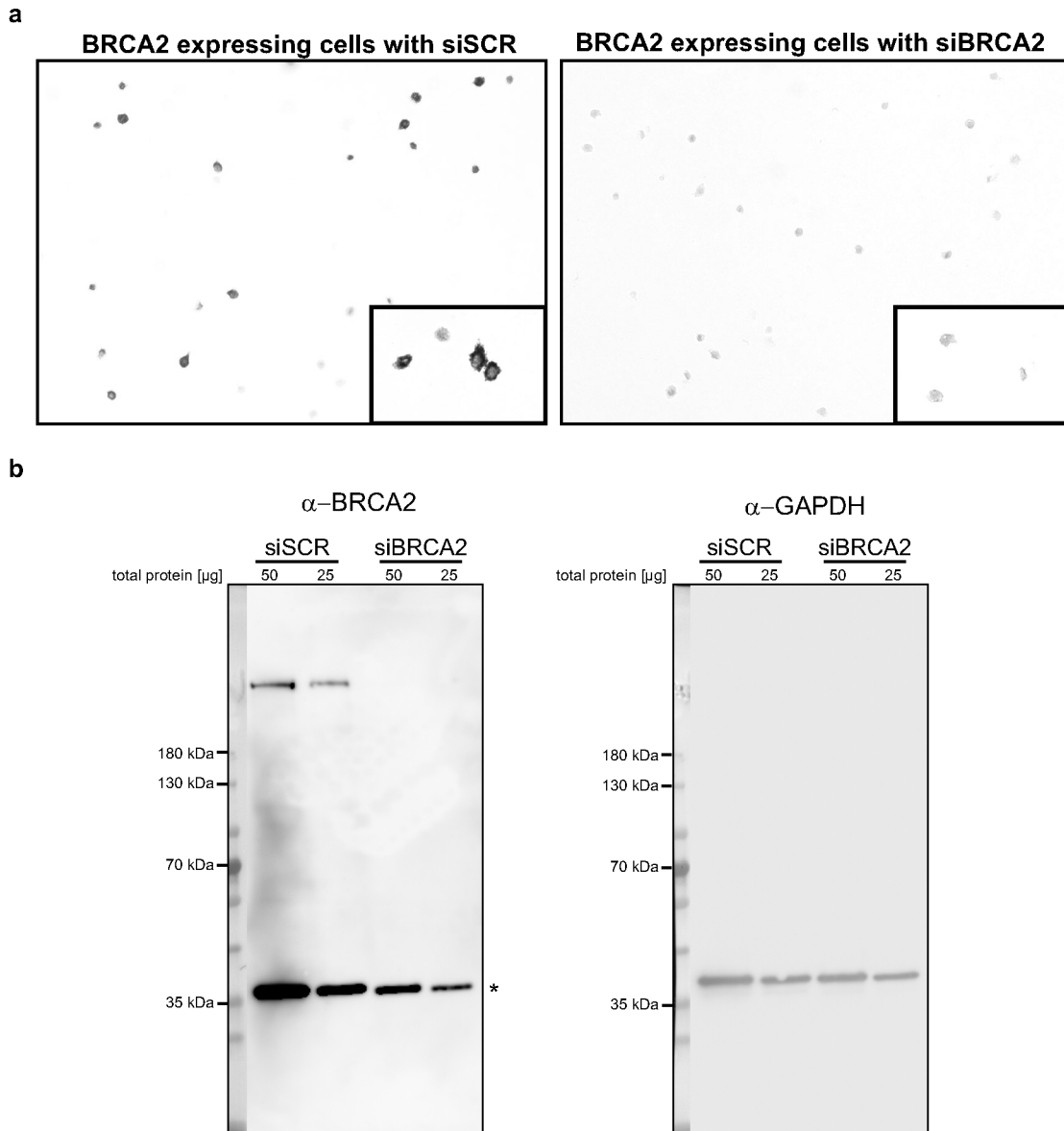
Supplemental table 2: Tumour-specific coding single nucleotide variants

Supplemental table 3: Tumour-specific coding small indels

Supplemental table 4: Recurrent tumour-specific copy number gains and losses

Supplemental table 5: Mutational signatures of the single tumours.

Supplemental Figure 1



Supplemental Fig. 1: Immunohistochemistry and Western blot analyses of BRCA2 expressing cells treated with BRCA2 siRNA confirm BRCA2 antibody specificity. The BRCA2 expressing prostate carcinoma cells line LNCAP was treated for 48 h with 43 nM BRCA2 siRNA (siBRCA2, 5'-CCAACUUUGUCCUUAACUA-3', Eurofins) or scrambled siRNA (siSCR, 5'-GCAGCUAUAUGAAUGUUGU-3', Eurofins), according to the Lipofectamine RNAiMAX protocols. **a**, In immunohistochemical analyses, LNCAP cells treated with the BRCA2 siRNA show hardly any BRCA2 protein expression. Treated cells were resuspended in HistoGel (#HG400012, ThermoScientific) following the manufacturers' protocols, then fixed with 4% formalin and embedded in paraffin. One μ m sections of these cell blocks were stained for BRCA2 (see method section in main text). Representative images at 100x and 400x (inlays) magnification were taken of the siSCR (left panel) and siBRCA2 (right panel) treated cells showing strong and no BRCA2 staining, respectively. **b**, In Western blot analyses, LNCAP cells treated with the BRCA2 siRNA do not display any discernible BRCA2 protein band at 380 kDa. Treated cells were lysed in IP-lysis buffer (50 mM Hepes, pH7.5; 150 mM NaCl; 10% glycerol; 1% Triton-X-100; 1.5 mM MgCl₂; 1 mM EDTA). 25 and 50 μ g protein were loaded onto 4-20% gradient gels (#456-1083, BioRad) and blotted onto nitrocellulose membranes (#170-4159, BioRad). Membranes were incubated with BRCA2 (#10741) or GAPDH (#2118) antibodies from Cell Signaling at a 1:1000 dilution o/n at 4°C, followed by an incubation with a HRP-conjugated, rabbit secondary antibody (#NA934, GE Healthcare) at 1:3000 dilution for 1 h at RT. Proteins were detected with Dura Extended Duration substrate (#34075, Thermo Fisher Scientific) and visualized using the ImageQuant LAS 4000 detection system (GE Healthcare). Images of the uncut BRCA2- (left panel) and GAPDH- (right panel) stained membranes are shown. *is a non-specific band at 35 kDa detected with the BRCA2 antibody.

Supplemental Table 1: Whole-exome sequencing coverage statistics*

Patient ID	Sample Type	Coverage	Mapped Reads %	Duplicates%	Properly Paired%	Insert Size (Median)
C000-2SXE39	CONTROL	224,98	99,99	33,68	97,55	149
C000-2SXE39	TUMOR	278,03	99,99	24,65	96,87	162
C000-648QQH	CONTROL	78,02	99,99	68,38	96,97	132
C000-648QQH	TUMOR	71,98	99,99	67,19	97,27	134
C000-6KNGDL	CONTROL	108,14	99,98	55,07	96,58	137
C000-6KNGDL	TUMOR	178,62	99,98	29,92	91,94	135
C000-AX1WLG	CONTROL	257,71	99,99	28,49	97,93	161
C000-AX1WLG	TUMOR	243,26	99,99	23,6	97,78	159
C000-CFLP5N	CONTROL	34,15	99,97	80,74	95,23	120
C000-CFLP5N	TUMOR	35,65	99,98	81,84	96,05	120
C000-ESMKCH	CONTROL	37,32	99,98	78,11	94,86	123
C000-ESMKCH	TUMOR	54,64	99,99	72,62	96,74	123
C000-EXKBCS	CONTROL	221,06	99,98	41,05	96,97	152
C000-EXKBCS	TUMOR	176,44	99,99	45,4	96,91	149
C000-HZGGV2	CONTROL	113,57	99,98	56,14	95,87	132
C000-HZGGV2	TUMOR	145,67	99,99	43,49	94,66	126
C000-JMUDMJ	CONTROL	170,8	99,99	45,81	97,68	148
C000-JMUDMJ	TUMOR	169,39	99,99	45,27	97,25	147
C000-RCWM1J	CONTROL	77,21	99,96	58,57	93,74	133
C000-RCWM1J	TUMOR	185,73	99,98	24,87	90,85	137
C000-S5ZHWR	CONTROL	144,49	99,98	47,84	97,31	144
C000-S5ZHWR	TUMOR	237,27	99,99	35,62	97,04	151
C000-W91YB6	CONTROL	155,08	99,99	40,2	97,87	149
C000-W91YB6	TUMOR	287,49	99,99	34,14	97,72	146
C000-X88E7Z	CONTROL	162,19	99,98	33,21	97,58	149
C000-X88E7Z	TUMOR	488,43	99,99	48,13	96,17	140
C000-ZZP8UU	CONTROL	87,77	99,99	65,91	96,17	125
C000-ZZP8UU	TUMOR	60,2	99,98	78,65	95,36	125
C000-XWCDE5*	CONTROL	4,76	99,95	91,63	97,29	110
C000-XWCDE5*	TUMOR	19,05	99,97	88,5	97,06	115
C000-DZCVJ8*	CONTROL	19,49	99,96	88,44	95,27	117
C000-DZCVJ8*	TUMOR	14,61	99,97	91,27	97,59	116

*samples with a median coverage of < 20X. Those were excluded from the analysis.

X	4158623	T	A	TGGATTGCCA	AAGCCGCTAT	exonic	GPR82	nonsynonymous SNV	GPR82:ENST00000302548.4:exon3:c.T344A:p.L115K
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X	68749626	G	T	CAACAAGCCC	CCCTGCTGCC	exonic	FAM155B	nonsynonymous SNV	FAM155B:ENST00000252338.4:exon3:c.G1246T:p.A416S,
X	107977066	G	T	CCCTCCCCAG	AACTTTCCA	exonic	IRS4	nonsynonymous SNV	IRS4:ENST00000372129.2:exon1:c.C2508A:p.L837M,
X	107977067	G	T	CCCTCCCCAG	AACTTTCCA	exonic	IRS4	nonsynonymous SNV	IRS4:ENST00000372129.2:exon1:c.C2508A:p.L836L,
X	144904814	C	A	TACAATGAAT	CTGCTCTCAA	exonic	SLITRK2	nonsynonymous SNV	SLITRK2:ENST00000335565.4:exon5:c.C871A:p.P291T,SLITRK2:ENST00000413937.2:exon3:c.C871A:p.P291T,SLITRK2:ENST00000447897.2:exon2:c.C871A:p.P291T,SLITRK2:ENST00000434188.2:exon3:c.C871A:p.P291T,SLITRK2:ENST00000370490.1:exon1:c.C871A:p.P291T,SLITRK2:ENST0000042891
X	151900103	C	A	GTCTCCCTC	CATCAGATGC	exonic	MAGEA12	nonsynonymous SNV	MAGEA12:ENST00000393900.3:exon3:c.G698T:p.G233V,MAGEA12:ENST00000357916.4:exon2:c.G698T:p.G233V,MAGEA12:ENST00000393869.3:exon3:c.G698T:p.G233V,

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COGONOMY

Table with 5 columns: Chromosome, Position (Pg), Reference allele, Mutant allele, Sequence context, and ANNOVAR function. Contains genomic data for various chromosomes and positions.

Table with 3 columns: Mutation type, ANNOVAR output (transcript), and Cytosine/CGMCG. Contains detailed mutation information and associated genomic features.

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Chromosome	Position (hg1)	Reference	all Mutant allele	Sequence context	ANNOVAR function	Gene	Mutation type	ANNOVAR output (alternative transcripts)	Cytoband	COSMIC
2	152584365	G	C	GGAAGTTCTATTGTCATA	exonic	NEB	stopgain	NEB:ENST00000427231.2:exon5:c.134G>S45X;NEB:ENST00000397345.3:exon5:c.134G>S45X;NEB:ENST0000064884.1:exon3:c.134G>S45X;NEB:ENST00000303839.1:exon3:c.134G>S45X;NEB:ENST00000409198.1:exon5:c.134G>S45X;NEB:ENST00000172853.10:exon5:c.134G>S45X;ZSWIM2:ENST00000295131.2:exon4:c.G344A;p.R115Q,	2q23.3	-
2	187703836	C	T	AGTTTGAACACTGATGATCCC	exonic	ZSWIM2	nonsynonymous SNV	ZSWIM2:ENST00000295131.2:exon4:c.G344A;p.R115Q,	2q32.1	-
5	35800251	T	C	GCAGAAAAGGAAATTGCATTC	splicing	SPEF2	-	SPEF2:ENST00000356031.3:exon34:c.5010+2T>C;ENST00000440985.2:exon34:c.4995+2T>C;ENST00000506526.1:exon12:c.2629+2T>C;ENST00000303129.4:exon10:c.1401+2T>C;ENST00000513078.1:exon10:c.1625+2T>C)	5p13.2	-
6	25850052	C	A	ATATCTAAGAATTGATATAA	exonic	SLC17A3	nonsynonymous SNV	SLC17A3:ENST00000397060.4:exon10:c.G1252T;p.V418F,	6p22.2	-
6	30916386	C	T	TCACATATTTCCCTCACTCC	exonic	DPCR1	nonsynonymous SNV	DPCR1:ENST00000462448.1:exon2:c.C145T;p.P49S,	6p21.33	-
7	123152327	G	A	AGTATCAAAA.CATCATCACT	exonic	IQUB	nonsynonymous SNV	IQUB:ENST00000434450.1:exon2:c.C68T;p.A23V;IQUB:ENST00000324698.6:exon2:c.C68T;p.A23V;IQUB:ENST00000405057.1:exon2:c.C68T;p.A23V;IQUB:ENST00000466202.1:exon2:c.C68T;p.A23V;IQUB:ENST00000444508.1:exon2:c.C68T;p.A23V,	7q31.32	-
8	113668499	C	T	ATTGTAAGAT.CAAGCAAGGG	exonic	CSMD3	nonsynonymous SNV	CSMD3:ENST00000339701.3:exon5:c.G908A;p.G303E;CSMD3:ENST00000297405.5:exon18:c.G2888A;p.G963E;CSMD3:ENST00000343508.3:exon19:c.G2788A;p.G923E;CSMD3:ENST00000455883.2:exon17:c.G2576A;p.G859E;CSMD3:ENST00000352409.3:exon18:c.G2888A;p.G963E,	8q23.3	CSMD3:basechange=c.2888G>T;AAchange=p.G863V;issue=lung
11	8496269	C	T	ATAATTTTTT.TTTTTTCTC	exonic	STK33	nonsynonymous SNV	STK33:ENST00000396673.1:exon3:c.G184A;p.E62K;STK33:ENST00000447869.1:exon1:c.G184A;p.E62K;STK33:ENST00000431279.2:exon3:c.G184A;p.E62K;STK33:ENST00000534493.1:exon3:c.G61A;p.E21K;STK33:ENST00000396673.1:exon3:c.G184A;p.E62K;STK33:ENST00000418597.1:exon2:c.G61A;p.E21K;STK33:ENST00000457885.2:exon2:c.G184A;p.E62K;STK33:ENST00000524760.1:exon1:c.G34A;p.E12K;STK33:ENST00000422559.2:exon3:c.G61A;p.E21K;STK33:ENST00000315294.1:exon3:c.G184A;p.E62K,	11p15.4	-
14	24780924	G	A	GATGGAGAAACGGTCCCGA	exonic	LTB4R2	nonsynonymous SNV	LTB4R2:ENST00000533293.1:exon2:c.G1054A;p.D352N;LTB4R2:ENST00000528054.1:exon1:c.G1147A;p.D383N;LTB	14q12	-
19	10480547	G	T	GTGAACCCC.TCTCTACTAA	exonic	TYK2	nonsynonymous SNV	4R2:ENST00000543919.1:exon2:c.G1054A;p.D352N,	19p13.2	-
22	26224742	G	T	TCTCTCCCTAATCCTTTTCC	splicing	MYO18B	nonsynonymous SNV	TYK2:ENST00000530829.1:exon4:c.C200A;p.T67K,	22q12.1	-
								1G>T;ENST00000407587.2:exon15:c.2787-1G>T;ENST00000335473.7:exon15:c.2787-1G>T;ENST00000407587.2:exon15:c.2787-1G>T;ENST00000539302.1:exon13:c.2642-1G>T)		

Table with 10 columns: Chromosome Position (qRefSeq), Mutant Alleles, Sequence context, ANNOVAR Func, Mutation type, ANNOVAR output (alternative transcripts), Cytoband, COBNC. Contains genomic data for chromosome 10.

Table with 2 columns: Cytoband, COBNC. Contains cytoband and COBNC identifiers for each genomic position.

sechange<4900>AAchangeP.R147QIssueLarge_inteliv

AAchangeP.Q205EIssueLung

AAchangeP.P220TIssueLung&RES1_EN010000381486_basechange<556C>AAchangeP.P220T.IssueLun

FIG-IT_AAchangeP.E120'IssueMedonmeter

EB9'IssueIn

egje: 16720>TAAChangep.R5MC.IssueEndometriu

300537824.baschangen-c.07150>T.AAChangep.V223F.IssueInge_Steine

3>T.AAChangep.C1381F.IssueWay

PRD_ENG1000034816.baschangen-c.4102C>TAAChangep.R109C.IssueBreast.prostate.endometriu&PFRD.baschangen-c.592C>TAAChangep.R109C.IssueBreast.prostate.endometriu&PFRD_ENG10000381196.baschangen-c.592C>TAAChangep.R109C.IssueBreast.prostate.endometriu

<24660>TAAChangep.G619V.IssueIn

plm-12900-1-Altchange-D4030basechange_000000

p-11870-basechange_000000

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021120000000000000

021120000000000000

021120000000000000

02050000

1950

910750

AAAHgngy 01099baaangMCCiEbaaazqum 12410AAAHgngy 04478baaang

2260baaang_thead

2baaang

1102baaangthead

01149baaangthead

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Table with 4 columns: Index, Gene Symbol, Gene Name, and Description. The index ranges from 2 to 54. Gene symbols include various mitochondrial genes like ATP5B, ATP5C1, ATP5A1, etc. The descriptions provide accession numbers and coordinates for each gene.

age_intestine

ng&RHBG.basechange=c.285C>T.AAchange=p.L95L.tissue=ovary.lung&RHBG_ENST00000368246.basechange=c.285C>T.AAchange=p.L95L.tissue=ovary.lung
ge_intestine&NTRK1.basechange=c.2074C>T.AAchange=p.R692C.tissue=large_intestine

ig&OR2L13.basechange=c.793C>T.AAchange=p.R265C.tissue=lung

Sbasechange<1532C>T,AAchange<p.S511F,issue=large_intestine

mploid_issue

dometrium&FAT4,basechange<6022C>T,AAchange<p.R2008W,issue=endometrium

lung

xus_system

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t

pe=c.662C>T;AAchange=p.S221F.tissue=endometrium

.jpg&CSMD1_ENST00000537824.basechange=c.7877C>T;AAchange=p.S2626F.tissue=lung

3

change: 113C-AAchange:p.538Y issue:pancreas

IST0000294064.basechange=c.364C>T,AAchange=p.P122S,tissue=large_intestine

ct

lvary_gland&ENOX1.basechange=c.724C>A,AAchange=p.R242R,tissue=lung&ENOX1.basechange=c.724C>T,AAchange=p.R242W,tissue=salivary_gland&ENOX1_ENST00000261488.basechange=c.724C>A,AAchange=p.R242R,tissue=lung

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_tract&TP53_ENST00000269305.basechange=c.1001G>T.AAchange.p.G334V.tissue=lung&TP53.basechange=c.1001G>C.AAchange.p.G334A.tissue=lung&TP53_ENST00000269305.basechange=c.1001G>C.AAchange.p.G334A.tissue=lung

trium

east&MYH14.basechange=c.982C>T.AAchange.p.R328W.tissue=breast

kidney&ZSCAN18.basechange=c.880G>A.AAchange=p.E294K.tissue=kidney

ometrium&FLCB1.basechange=c.1229C>T.AAchange=p.S410L.tissue=endometrium
00378641.basechange=c.2753C>T.AAchange=p.S918L.tissue=lung

ICK.basechange=c.310G>T.AAchange=p.E104*
tissue=lung

am

ometrium&TAF1.basechange=c.3505C>T.AAchange=p.R1169C.tissue=endometrium

...A... ...B... ...C... ...D... ...E... ...F... ...G... ...H... ...I... ...J... ...K... ...L... ...M... ...N... ...O... ...P... ...Q... ...R... ...S... ...T... ...U... ...V... ...W... ...X... ...Y... ...Z...

...A... ...B... ...C... ...D... ...E... ...F... ...G... ...H... ...I... ...J... ...K... ...L... ...M... ...N... ...O... ...P... ...Q... ...R... ...S... ...T... ...U... ...V... ...W... ...X... ...Y... ...Z...

...A... ...B... ...C... ...D... ...E... ...F... ...G... ...H... ...I... ...J... ...K... ...L... ...M... ...N... ...O... ...P... ...Q... ...R... ...S... ...T... ...U... ...V... ...W... ...X... ...Y... ...Z...

...A... ...B... ...C... ...D... ...E... ...F... ...G... ...H... ...I... ...J... ...K... ...L... ...M... ...N... ...O... ...P... ...Q... ...R... ...S... ...T... ...U... ...V... ...W... ...X... ...Y... ...Z...

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...A... ...B... ...C... ...D... ...E... ...F... ...G... ...H... ...I... ...J... ...K... ...L... ...M... ...N... ...O... ...P... ...Q... ...R... ...S... ...T... ...U... ...V... ...W... ...X... ...Y... ...Z...

Chromosomes/Position (hg)	Reference Allele	Mutant Allele	Sequence Context	ANNOVAR	Gene	Mutation Type	ANNOVAR output (alternative transcripts)
1	1355494	C	TCCGGGGGC_CACCCGGCCC	exonic	ANKRD65	nonsynonymous SNV	ANKRD65:ENST00000357107.1 exon3:c.688A>G;D330N,
1	1626364	C	CGACTTCGT_CCCCTCCGCTG	exonic	SPFN	nonsynonymous SNV	SPFN:ENST00000379759.3 exon11:c.C1029G>T;p.394T1L,
1	1952456	T	GCTGAGGGT_ATATCGTCAT	exonic	UBR4	stopgain	UBR4:ENST0000037256.3 exon2:c.C68T>T;p.148R4;ENST0000375217.2 exon2:c.C68T>T;Q229X,UBR4:ENST0000037526.2 exon2:c.C68T>T;Q229X,
1	1556727	C	GAATACGCT_AGGGAGCCCA	exonic	USP24	nonsynonymous SNV	USP24:ENST00000294383.6 exon4:c.G5143C;p.E1715Q;USP24:ENST00000407756.1 exon4:c.Q4663C;p.E1555Q,
1	16272229	G	CAACATGCTCA_TGAGGACAT	exonic	DDR2	nonsynonymous SNV	DDR2:ENST00000446985.1 exon2:c.G127C;p.D43H;DDR2:ENST00000367922.3 exon5:c.G127C;p.D43H;DDR2:ENST00000367921.3 exon4:c.G127C;p.D43H,
1	16803472	T	CGACGACAT_CGTTTGACCC	exonic	DFAF6	nonsynonymous SNV	DFAF6:ENST0000031283.6 exon16:c.C231T;p.P771S;DFAF6:ENST0000042587.2 exon18:c.C2491T;p.P831S;DFAF6:ENST0000036784.3 exon17:c.C2371T;p.P791S;DFAF6:ENST0000036784.3 exon19:c.C2584T;p.P862S,
1	24227125	A	TTTGATGTC_GATCCGGGCG	exonic	PLD5	nonsynonymous SNV	PLD5:ENST0000036534.2 exon4:c.T108T;p.L363V;PLD5:ENST0000427465.1 exon3:c.T19G1G;p.L30V;PLD5:ENST0000042994.2 exon2:c.T181G;p.L271V,
1	24229767	T	GATGCATCG_GTTCTTTTCA	exonic	PLD6	nonsynonymous SNV	PLD6:ENST0000036534.2 exon4:c.T108T;p.L363V;PLD6:ENST0000427465.1 exon3:c.T19G1G;p.L30V;PLD6:ENST000042994.2 exon2:c.T181G;p.L271V,
2	2538336	G	TCCCTTGCG_TATTACGAG	exonic	EFR3B	nonsynonymous SNV	EFR3B:ENST0000041432.3 exon15:c.G1312A;p.V43L;EFR3B:ENST00000264719.5 exon5:c.G94A>P;p.V317L;EFR3B:ENST00000405108.1 exon10:c.G868A;p.V290L;EFR3B:ENST00000402191.1 exon13:c.G1207A;p.V403L;EFR3B:ENST00000403714.2 exon13:c.G1312A;p.V4
2	54120833	A	AAACGCTCG_GATTTAACAAT	exonic	PSME4	nonsynonymous SNV	PSME4:ENST0000041748.2 exon15:c.C1448T;p.V483L;PSME4:ENST00000404125.1 exon35:c.C4015T;p.P1339S,
2	54120834	T	AAACGCTCG_GATTTAACAAT	exonic	PSME4	nonsynonymous SNV	PSME4:ENST0000041748.2 exon15:c.C1447T;p.P483S;PSME4:ENST00000404125.1 exon35:c.C4015T;p.P1339S,
2	19730279	T	TGTCCTTCT_GAGATCGCT	exonic	ANKRD36	stopgain	ANKRD36:ENST0000041531.2 exon2:c.C67A>T;p.M914K;ANKRD36:ENST00000420699.2 exon2:c.C67A>T;p.R22XK,
2	98887171	A	CCAATACAGT_CCTTGAGAAC	exonic	VWA3B	nonsynonymous SNV	VWA3B:ENST00000477737.1 exon22:c.A276G>C;p.Y957F,
2	11659832	T	GCAGCAAGA_TCAATTCCAA	exonic	DPP10	nonsynonymous SNV	DPP10:ENST00000409163.1 exon26:c.T2039G;p.V808G;DPP10:ENST00000410059.1 exon5:c.T2189G;p.V730G;DPP10:ENST000004093147.2 exon25:c.T2201G;p.V734G,
2	13491363	G	GATCGTGAAT_AAACATGGGG	exonic	NCKAP5	nonsynonymous SNV	NCKAP5:ENST00000405974.3 exon11:c.C1408G;p.H470D;NCKAP5:ENST00000402131.1 exon25:c.C1408G;p.H470D;NCKAP5:ENST00000371721.6 exon15:c.C5365G;p.H1789D;NCKAP5:ENST00000409261.1 exon17:c.C5365G;p.H1789D,
2	16000969	T	CGATGAGCC_CGCTCAACCT	exonic	TANC1	nonsynonymous SNV	TANC1:ENST0000026363.6 exon7:c.T574G>C;p.I925Q,
2	16373863	T	ACGTCTGGT_GGAGCTTAAAC	exonic	ITC21B	nonsynonymous SNV	ITC21B:ENST00000203344.2 exon8:c.T799A;p.S267F,
2	167133576	G	TCCACATCGT_CCTCATCCAC	exonic	SCN8A	nonsynonymous SNV	SCN8A:ENST0000037587.4 exon16:c.AE2794C;p.T832P;SCN8A:ENST00000409435.1 exon15:c.AE2794C;p.T832P;SCN8A:ENST00000303354.6 exon16:c.AE2794C;p.T832P;SCN8A:ENST0000040672.1 exon16:c.AE2794C;p.T832P,
2	17124362	C	CTATGACCTG_CATCATCATC	exonic	MYO3B	nonsynonymous SNV	MYO3B:ENST00000409044.3 exon14:c.C1451T;p.A484V;MYO3B:ENST00000408978.4 exon14:c.C1451T;p.A484V;MYO3B:ENST00000317935.3 exon14:c.C1451T;p.A484V;MYO3B:ENST00000334231.3 exon14:c.C1478T;p.A493V;MYO3B:ENST00000484338.2 exon14:c.C14
2	17834352	G	GTGATTTAA_GATTTGACCC	exonic	AGPS	nonsynonymous SNV	AGPS:ENST00000284167.4 exon4:c.G1369A;p.G457R,
2	17854353	G	TAGTTTTAA_GATTACCCCA	exonic	AGPS	nonsynonymous SNV	AGPS:ENST00000284167.4 exon4:c.G1370A;p.G457E,
2	22863332	A	TGTCTAGCT_GAGCCGCGCG	exonic	FZD5	nonsynonymous SNV	FZD5:ENST00000295417.3 exon2:c.C1320G>T;p.H44M,
2	21059938	C	ATGAGCCCTG_ATTTAATCTT	exonic	MAP2	nonsynonymous SNV	MAP2:ENST00000360351.4 exon11:c.G2144A;p.G171E;MAP2:ENST00000447185.1 exon4:c.G2132A;p.G711E,
2	21058106	T	ATTACAAGA_GAGACACCCA	exonic	MAP2	nonsynonymous SNV	MAP2:ENST00000360351.4 exon11:c.G2142A;p.D1374E;MAP2:ENST00000447185.1 exon4:c.T4110A;p.D1370E,
2	21897781	A	TACCATGAGT_ACTCCATCC	exonic	XRCO5	nonsynonymous SNV	XRCO5:ENST00000417391.1 exon3:c.A25T>N;XRCO5:ENST00000392133.1 exon3:c.A64T>N;XRCO5:ENST00000392132.1 exon2:c.A64T>N;XRCO5:ENST00000392131.1 exon2:c.A64T>N;XRCO5:ENST00000392130.1 exon2:c.A64T>N;XRCO5:ENST00000392129.1 exon2:c.A64T>N;XRCO5:ENST00000392128.1 exon2:c.A64T>N;XRCO5:ENST00000392127.1 exon2:c.A64T>N;XRCO5:ENST00000392126.1 exon2:c.A64T>N;XRCO5:ENST00000392125.1 exon2:c.A64T>N;XRCO5:ENST00000392124.1 exon2:c.A64T>N;XRCO5:ENST00000392123.1 exon2:c.A64T>N;XRCO5:ENST00000392122.1 exon2:c.A64T>N;XRCO5:ENST00000392121.1 exon2:c.A64T>N;XRCO5:ENST00000392120.1 exon2:c.A64T>N;XRCO5:ENST00000392119.1 exon2:c.A64T>N;XRCO5:ENST00000392118.1 exon2:c.A64T>N;XRCO5:ENST00000392117.1 exon2:c.A64T>N;XRCO5:ENST00000392116.1 exon2:c.A64T>N;XRCO5:ENST00000392115.1 exon2:c.A64T>N;XRCO5:ENST00000392114.1 exon2:c.A64T>N;XRCO5:ENST00000392113.1 exon2:c.A64T>N;XRCO5:ENST00000392112.1 exon2:c.A64T>N;XRCO5:ENST00000392111.1 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Chromosome	Position (hg)	Reference	Mutant allele	Sequence context	ANNOVAR ft	Gene	Mutation type	ANNOVAR output (alternative transcripts)
1	152882979	C	T	GCTGGAGCTC,CACAGCAGCA	exonic	IVL	nonsynonymous SNV	IVL:ENST00000368764.3:exon2:c.C706T;p.P236S,IVL:ENST00000392667.2:exon3:c.C268T;p.P90S,
1	168510311	G	A	TCTCACCAC,TTGGTTGG	exonic	XL2	nonsynonymous SNV	XL2:ENST00000367819.2:exon3:c.C224T;p.T75M,
2	167141144	C	T	GTTACTGCTG,GTGCTCCTG	exonic	SCN9A	nonsynonymous SNV	SCN9A:ENST00000409672.1:exon12:c.G1793A;p.R598H,SCN9A:ENST00000454569.1:exon9:c.G1388A;p.R463H,SCN9A:ENST00000409435.1:exon11:c.G1793A;p.R598H,SCN9A:ENST00000375387.4:exon12:c.G1796A;p.R599H,SCN9A:ENST000004
3	119198789	G	C	ATATATCTTT,ACAATGTGAC	exonic	POGLUT1	stoploss	POGLUT1:ENST00000486607.1:exon5:c.G491C;p.X184S,
3	16984623	A	G	GGCGAAACCG,GGAGACAA	exonic	SEC62	nonsynonymous SNV	SEC62:ENST00000337002.4:exon1:c.A13G;p.R5G,SEC62:ENST00000469515.2:exon1:c.A13G;p.R5G,SEC62:ENST00000487708.1:exon1:c.A13G;p.R5G,SEC62:ENST00000460513.1:exon1:c.A13G;p
6	87726137	G	A	ATTAGATGCC,AGAGCATACT	exonic	HTR1E	nonsynonymous SNV	HTR1E:ENST00000305344.5:exon2:c.G1085A;p.R362Q,
7	122221277	C	T	CCAGTGCTTT,TTGTAAGAGT	exonic	CADPS2	nonsynonymous SNV	CADPS2:ENST00000412584.2:exon7:c.G1291A;p.E431K,CADPS2:ENST00000313070.7:exon7:c.G1291A;p.E431K,CADPS2:ENST00000449022.2:exon7:c.G1291A;p.E431K,
7	137266651	G	C	GGCTGAAGTA,TTATTGAAAA	exonic	DGKI	nonsynonymous SNV	DGKI:ENST00000453654.2:exon15:c.C687G;p.N229K,DGKI:ENST00000446122.1:exon15:c.C1587G;p.N529K,DGKI:ENST00000424189.2:exon15:c.C1587G;p.N529K,DGKI:ENST00000288490.5:exon15:c.C1587G;p.N529K,
7	150217958	C	T	AAGTTTTATT,TTCTAATTT	exonic	GIMAP7	nonsynonymous SNV	GIMAP7:ENST00000313543.4:exon2:c.C896T;p.S299F,
7	150217961	C	T	TTTTATTCTT,CTAATTACT	exonic	GIMAP7	nonsynonymous SNV	GIMAP7:ENST00000313543.4:exon2:c.C899T;p.S300F,
8	110527432	C	G	TTCCACACTT,AACTTTTGA	exonic	PKHD1L1	nonsynonymous SNV	PKHD1L1:ENST00000378402.5:exon72:c.C11587G;p.Q3863E,PKHD1L1:ENST00000526472.1:exon18:c.C2371G;p.Q791E,
11	64757197	G	C	TGCACGTGCA,GGTCCGGCTC	exonic	BATF2	nonsynonymous SNV	BATF2:ENST00000534177.1:exon3:c.C226G;p.L76V,BATF2:ENST00000527716.1:exon2:c.C157G;p.L53V,BATF2:ENST00000301887.4:exon3:c.C229G;p.L77V,
12	11138642	G	A	TGAGTCGAAT,CAAGATATAT	exonic	TAS2R50	nonsynonymous SNV	TAS2R50:ENST00000506688.1:exon1:c.C818T;p.A273V,
12	50045835	C	T	CATGCCCTCA,TCAGCTCTCG	exonic	FMNL3	nonsynonymous SNV	FMNL3:ENST00000352151.5:exon13:c.G1331A;p.S444N,FMNL3:ENST00000335154.5:exon14:c.G1484A;p.S495N,FMNL3:ENST00000550488.1:exon14:c.G1484A;p.S495N,FMNL3:ENST00000293590.5:exon14:c.G1484A;p.S495N,
12	85441073	G	A	GAAAAATGTA,ACAGTCTTTT	exonic	LRRIQ1	nonsynonymous SNV	LRRIQ1:ENST00000393217.2:exon6:c.G503A;p.R168K,
14	101347482	C	T	GGGTTCTTGA,GCAGGGCAGG	exonic	RTL1	nonsynonymous SNV	RTL1:ENST00000544062.1:exon1:c.G364A;p.R1215H,
15	40253993	G	A	ACGCGAGAAC,TCAGTATTCT	exonic	EIF2AK4	nonsynonymous SNV	EIF2AK4:ENST00000382727.2:exon7:c.G752A;p.R251H,EIF2AK4:ENST00000263791.5:exon7:c.G752A;p.R251H,EIF2AK4:ENST00000559624.1:exon7:c.G752A;p.R251H,
16	4812577	T	C	TACTCACCTC,CTCTGGCCAC	exonic	ZNF500	nonsynonymous SNV	ZNF500:ENST00000545009.1:exon3:c.A595G;p.R199G,ZNF500:ENST00000219478.6:exon3:c.A595G;p.R199G,
17	39254126	C	T	CAGCTGGACA,ACAGCAGCTG	exonic	KRTAP4-8	nonsynonymous SNV	KRTAP4-8:ENST0000033822.4:exon1:c.G211A;p.V71M,
20	18440840	G	C	CCATCAGGCA,AGTAATAGGT	exonic	DZANK1	nonsynonymous SNV	DZANK1:ENST00000608192.1:exon3:c.C220G;p.L74V,DZANK1:ENST00000262547.5:exon3:c.C220G;p.L74V,DZANK1:ENST00000329494.5:exon3:c.C220G;p.L74V,DZANK1:ENST00000609267.1:exon3:c.C220G;p.L74V,DZANK1:ENST00000377630.5

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15	64017635	C	ATTGTTTITAGATCACA	exonic	HEB_C1
15	66791804	G	TTCTTTCAG_GGCTGGTCT	exonic	RPL4
15	77795549	A	CTTCTTCAAGAGCTCCGAGA	exonic	HMG20A
15	79750488	C	CACAGCTCTCCAGCGAAG	exonic	BLA
15	81046666	C	GAGGCTCTCC_CCGAGCGTG	exonic	ABHD17C
15	83438603	C	GTTTGCTACTACGAATTA	exonic	FSO2
15	83521308	C	TGTTCTCTTTGAATTTGGT	exonic	SLC12A4
15	89395189	G	TGCGCTCTAAAGTCCACAC	exonic	ACAN
15	90039699	T	CGAGGAGGTC_GAAGAAAGT	exonic	ZNF774
15	91347415	G	FATGGAAACA_AAAATCTCCAG	exonic	BLA
15	94422310	G	AAAGATTAAG_CAAAGATCTT	exonic	MCR22
15	26157202	C	CCCAAGGCAC_GAGGAAAGCT	exonic	SRRM2
15	11034654	C	CGCTTTGCGG_CAGCGCAGC	exonic	MNF25
15	3434278	C	CACAGGCTCTTCTCACTTC	exonic	ZSCAN3L
15	3591889	C	COTGGCCGCG_AGTCCCGCAT	exonic	NLRC3
15	4539722	C	CGGCTGCTGCTTTGATGCG	exonic	SLC14
15	21008782	T	ATGTTGAGGTATTGGCGGAA	exonic	DNAH3
15	23464533	T	ACTTGATCCTTTATTTTAT	exonic	PALB2
15	24246523	T	TGTTCTGTCAA_AAGCTGGTCA	exonic	RHCP41P17
15	55683700	C	GAGGAGGCTTAATAAAAGT	exonic	CESSA
15	66838906	T	CCCGCGACTT_CCGCGCCAAA	exonic	TK2
15	69232947	C	GAGCGGCTCT_GAAGTGGCA	exonic	SLC12A4
15	69664472	C	GTTGGTGGCCAA_CCGGCGACC	exonic	DDX28
15	71610150	G	ACATGAGCTC_GATTGGGTTG	exonic	TAT
15	88496959	G	CCGCTCCCGAA_GAGGAGTGA	exonic	ZNF469
15	89329497	G	GGAGATTCAT_AAGCTTCCCTG	exonic	ZNF469
15	89795707	C	CGACTGCTCTT_AAGCTTACC	exonic	ZNF276
15	90026290	C	TTCCTGCTCTT_CCTGGGCTCA	exonic	DEFB
15	90158617	C	CACCTTTCGA_GATGAGGCTCC	exonic	DDX28
15	986762	C	CGGCTACTCTT_GATGATCTTC	exonic	ABR_NEST000057142.1
15	1939605	T	ACAACTTTGAATTTCCCAAG	exonic	DPH1
15	31815233	C	GAACCGACTT_GTCAGACTCC	exonic	OR3A2
15	31956688	G	GTATAGGCGG_TGTGAGTTAT	exonic	OR3A1
15	36287117	G	TCCAGCCTCTT_AAGCTTGGCT	exonic	ATP2A3
15	4486244	G	TGCGGCTCCTT_AAGTCTGGTG	exonic	SMN2
15	4883766	G	GTCGCTGCTT_GAAGCTGACC	exonic	CAMTA2
15	7098610	A	GTCTTGGTGG_GGCCAAGCTC	exonic	DLG4
15	9295868	C	AACTCACTGA_AACTATGATC	exonic	NEURL4
15	12504110	A	AATCGCGAGC_GAAGGAGGAC	exonic	C3P1
15	17343026	C	AGAACCTTCTT_CCTCTTGGAC	exonic	SHBG
15	17877010	T	GGAGCGGCTT_AGGAGGAGT	exonic	TPS38B
15	7678188	T	TACTGGTCTT_CAAATGACAG	exonic	DNAH2
15	8215376	C	CCAGTCCGCTT_CTCGACGACC	exonic	ARRHG15
15	8434562	C	FATCGGCTCC_AAAGGCTGGC	exonic	MYOC
15	12639462	A	AGCTGCTCTG_GAGCAACAAC	exonic	ACO3538_1.MYOC
15	12647507	C	GATGTCAAAAT_CTTGGGTGCT	exonic	ACO3538_1.MYOC
15	12656433	C	CCAGGCTTCT_GAAGATGAGC	exonic	ACO3538_1.MYOC
15	15973811	G	AGTGAACCTG_AATGATGGTG	exonic	NCO11
15	16395180	C	GCCGCCGCGC_CCGCCTGCTC	exonic	FAM21A
15	16398510	C	CCAACTTGAA_TGGTCTTGGT	exonic	NLRC3
15	18881189	C	GGAAGCTGCTT_AAGTCTGGTC	exonic	FAM33G
15	26715283	C	AAGCAAGTGT_CCAAGCTGGC	exonic	SARM1
15	27925625	C	TCAGGCTCTT_AAGCTGGTCC	exonic	TLCE3
15	29183993	G	GTGCGATTG_AACCCACCTC	exonic	ATAD5
15	30274665	G	GATGATTTT_AAAGATGACT	exonic	SUF2F2
15	30521255	C	GCGCTTCTT_CTCACAAATG	exonic	RHNOB3
15	30964090	T	TCTCAAGCTT_ACAACTAGT	exonic	ZNF207
15	34894142	G	GCCCGGCGCG_TGGCGTCCGC	exonic	MRM1
15	35585181	A	GCGCAATTCG_CAGCACTGGC	exonic	SYNRG1
15	36731149	A	GCCCGGAGCG_GGGCGAGTGC	exonic	SRGN1
15	37439073	G	GTCAACCAAG_AAATGTTCAA	exonic	FXB120
15	37898919	A	CCCGCGCTCT_GAGTCTGGTC	exonic	STAT5A
15	40447783	C	CACTTCACTG_CAGCTGAGTC	exonic	STAT5A
15	41003806	C	TCGGCTCACC_CTCCTGACTG	exonic	GUC1
15	42862827	C	CCGGCTGCTT_CCGGCTGGCT	exonic	AOC3
15	44467163	C	CGCCGCGCTT_AGTGTTGGTG	exonic	WNT3
15	46509000	G	TTCGTGGACA_AAGCGACTCG	exonic	CDKSRAP3
15	47218648	A	TGTCCTAACT_CTCGATCTGT	exonic	BACAL21P3
15	47265449	T	CCGAAAGATG_AATCTCAACT	exonic	MK51
15	58330564	C	TTCCTCACTT_AAAGAGCTG	exonic	LPF32
15	60088536	T	TGTGTTCTCT_AACTAAAGAT	exonic	MED13
15	61903476	C	GCGAGCACTG_CCTTCAAGAT	exonic	FTL3
15	71231635	T	AGTGAATAT_CACCGACAAG	exonic	C17orf80
15	73734247	C	TGCGGCTGCTT_CTCGGGAGC	exonic	ITGB8
15	75266589	C	GATCGCCAGT_AAGCTCGTAT	exonic	RHBDF2
15	76183457	A	AGCGGCTAGT_GATGTTGGTG	exonic	AFM1D
15	76183458	A	CGCTCAGTAT_GATGTTGGTG	exonic	AFM1D
15	76183459	A	CGCAACATCA_CCTCTCTCTT	exonic	NM00017
15	76183460	A	CGCAGCACTG_AATTTGGGTT	exonic	RNF123
15	76822117	C	GTCTTGCTGG_CCTCTCGACG	exonic	SLC25A10
15	76848193	C	CGCCGCTAGT_TGCGTCTGTC	exonic	CDFR
15	79842762	C	GGTTTCTAGT_AAGCTGTACT	exonic	ALYRF
15	79918493	C	CGAGTCCACC_AAGTGTTTGAG	exonic	NOTUM
15	79918494	C	AAAGTCGACA_CTTCTCGAGT	exonic	SOX22
15	12802143	C	AAAGTCTCTT_CATGCTTTCT	exonic	PTNP2
15	12802144	C	AAAGTCTCTT_CATGCTTTCT	exonic	PTNP2
15	13232516	C	GGGAAAGTAT_AATTGAGTGT	exonic	PTRN1
15	13232517	C	TTTCAATCAI_AAGTACCATC	exonic	ROCK1
15	13232518	C	TTTCAATCAI_AAGTACCATC	exonic	GREB1L
15	13232519	C	TTTCAATCAI_AAGTACCATC	exonic	MI6
15	13232520	C	TTTCAATCAI_AAGTACCATC	exonic	LAMA3
15	13232521	C	TTTCAATCAI_AAGTACCATC	exonic	KLHL4
15	13232522	C	TTTCAATCAI_AAGTACCATC	exonic	KLHL4
15	13232523	C	TTTCAATCAI_AAGTACCATC	exonic	CCDC178
15	13232524	C	TTTCAATCAI_AAGTACCATC	exonic	ASXL3
15	13232525	C	TTTCAATCAI_AAGTACCATC	exonic	ASXL3
15	13232526	C	TTTCAATCAI_AAGTACCATC	exonic	TPS2
15	13232527	C	TTTCAATCAI_AAGTACCATC	exonic	TPS2
15	13232528	C	TTTCAATCAI_AAGTACCATC	exonic	KOR2L
15	13232529	C	TTTCAATCAI_AAGTACCATC	exonic	CEBE1
15	13232530	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232531	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232532	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232533	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232534	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232535	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232536	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232537	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232538	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232539	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232540	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232541	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232542	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232543	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
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15	13232545	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232546	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232547	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232548	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
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15	13232552	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
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15	13232563	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
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15	13232567	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232568	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
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15	13232570	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232571	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232572	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232573	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232574	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232575	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232576	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232577	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232578	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232579	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232580	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232581	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232582	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232583	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232584	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
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15	13232586	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232587	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232588	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232589	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232590	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232591	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232592	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232593	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232594	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232595	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232596	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232597	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232598	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232599	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236
15	13232600	C	TTTCAATCAI_AAGTACCATC	exonic	ZNF236

X	37545217	G	A	GGCGGGAGTAAATTCGGG	exonic	XX	nonsense	XX:ENST00000378616.3:exon1:c.G3A:p.M11.
X	41555743	G	A	CATCCCTTTCATCATCTAC	exonic	XX	nonsense	GPR34:ENST00000378158.5:exon3:c.G857A:p.R286Q.
X	47511482	C	T	GCTCTCACCTAGCAACATG	exonic	XX	nonsense	UXT:ENST0000035890.2:exon5:c.G454A:p.E152K.UXT:ENST00000333119.3:exon6:c.G418A:p.E140K.
X	48434879	G	C	ACGTCCCAAGACTATAATGG	exonic	XX	nonsense	RBM3:ENST00000354480.2:exon2:c.G318C:p.E106D.RBM3:ENST00000376755.1:exon4:c.G400C:p.D134H.RBM3:ENST00000430348.2:exon4:c.G318C:p.E106D.
X	48543884	C	T	TCGCCCCCAACCCCTTCTTC	exonic	XX	nonsense	WAS:ENST00000370701.4:exon3:c.C3327:p.T1111.WAS:ENST00000460772.1:exon5:c.C3327:p.T1111.
X	48565911	C	T	GAAGCGGGTCGTATTGAATG	exonic	XX	nonsense	SUV39H1:ENST00000337852.6:exon6:c.C12221:p.R408C.SUV39H1:ENST00000376887.3:exon6:c.C1189T:p.R397C.
X	4896765	C	T	GGCGCTGTCCGACGCTCACG	exonic	XX	nonsense	TFE3:ENST00000315869.7:exon3:c.G401A:p.R134Q.
X	49066604	C	T	AGCTCAACGCTGGTGGGGGG	exonic	XX	nonsense	CACNA1F:ENST00000376251.1:exon4:c.G4525A:p.D1509N.CACNA1F:ENST00000376255.2:exon40:c.G4720A:p.D1574N.CACNA1F:ENST00000323022.5:exon40:c.G4687A:p.D1563N.
X	53283848	C	T	CCGGTGGCTGGCCACACGAG	exonic	XX	nonsense	IQSEC2:ENST00000375365.2:exon4:c.G650A:p.R217Q.IQSEC2:ENST00000396435.3:exon4:c.G1265A:p.R422Q.IQSEC2:ENST00000375368.5:exon3:c.G1235A:p.R412Q.
X	62855285	G	A	TGAGGAAAGGAGCTCAGGG	exonic	XX	nonsense	ARHGEP9:ENST00000374878.7:exon7:c.C1351T:p.P451S.
X	67940227	G	A	GAGCATTGAGAGCGATGGG	exonic	XX	nonsense	STARDB8:ENST0000034599.3:exon8:c.C2011G:p.G817E.STARDB8:ENST00000252336.6:exon7:c.C1771G:p.Q591E.STARDB8:ENST00000374597.3:exon7:c.C1771G:p.Q591E.
X	67941564	C	T	CTCAATGCTCTAAGAAAGAT	exonic	XX	nonsense	STARDB8:ENST00000374599.3:exon10:c.C2435T:p.S812F.STARDB8:ENST00000252336.6:exon9:c.C2195T:p.S732F.STARDB8:ENST00000374597.3:exon9:c.C2195T:p.S732F.
X	69353912	C	T	CCGATAGTCAGGAGAAAGT	exonic	XX	nonsense	IGBP1:ENST00000356413.4:exon2:c.C1157:p.Q39X.IGBP1:ENST00000342206.6:exon1:c.C1157:p.Q39X.
X	69421915	G	A	TGGAAACAGGGTGGCTCCAA	exonic	XX	nonsense	DGA12L1:ENST00000333028.3:exon5:c.G47+1G.
X	70339188	G	A	CTTCCCTAAGAAAAACAA	exonic	XX	nonsense	MED1:ENST00000429213.1:exon2:c.20-1G(A).
X	73811873	G	C	AGTAGGCTCTAGTCGCTACT	exonic	XX	nonsense	RLIM:ENST00000332887.6:exon4:c.C1277G:p.S426X.RLIM:ENST00000349225.2:exon5:c.C1277G:p.S426X.
X	76837199	C	G	TTCTCTTTTTCCTTGACAA	exonic	XX	nonsense	ATRX:ENST00000373344.4:exon9:c.G3549C:p.E1183D.ATRX:ENST00000395603.3:exon8:c.G3435C:p.E1145D.
X	83129181	G	A	AATGGATCTATTGGAGTT	exonic	XX	nonsense	CYL1C:ENST00000329312.4:exon4:c.G1465A:p.D489N.
X	84526170	G	A	TGTATTTCAGTGTCCAGAT	exonic	XX	nonsense	ZNF711:ENST00000360700.4:exon10:c.G1769A:p.R587K.ZNF711:ENST00000542788.1:exon7:c.G1148A:p.R383K.ZNF711:ENST00000276123.3:exon10:c.G1622A:p.R541K.ZNF711:ENST00000395402.1:exon9:c.G1646A:p.R549K.ZNF711:ENST00000373165.3:exon9:c.G1622A:p.R541K.
X	86893982	C	T	TATTGGTGGAGTGGATGAAG	exonic	XX	nonsense	KLHL4:ENST0000037119.4:exon2:c.C17327:p.R579C.KLHL4:ENST0000037114.4:exon9:c.C17327:p.R579C.
X	102471224	G	A	ATAGCTGGAGAAATACGAG	exonic	XX	nonsense	BEX4:ENST00000372891.3:exon2:c.G143A:p.G48E.BEX4:ENST00000372895.5:exon3:c.G143A:p.G48E.
X	104463988	G	A	ATAGGCGAGGAGCTGGACAG	exonic	XX	nonsense	TEX13A:ENST00000372575.1:exon3:c.C890T:p.S297F.TEX13A:ENST00000372578.3:exon3:c.C890T:p.S297F.
X	10639610	A	C	CTGCTGTGACGGATAAATC	exonic	XX	nonsense	NUP200CL:ENST00000374965.4:exon7:c.T422G:p.L141R.
X	106891314	G	A	GGCAAAGTGGGATTGATAAA	exonic	XX	nonsense	GUCY2F:ENST00000218006.2:exon6:c.C15537:p.P516L.
X	11202236	G	A	TGTTTGTCTGATTCAGAT	exonic	XX	nonsense	AMOT:ENST00000371962.1:exon10:c.C2460T:p.P817L.AMOT:ENST00000524145.1:exon11:c.C3146T:p.P1049L.AMOT:ENST00000371969.3:exon10:c.C3146T:p.P1049L.
X	11442654	C	T	AGGGGGGATGGCGATGCG	exonic	XX	nonsense	RBML3:ENST00000424775.3:exon1:c.C2830T:p.S977L.
X	117033075	T	A	CCACATAGATTATTTTCCA	exonic	XX	nonsense	KLHL13:ENST00000371882.1:exon7:c.A1611T:p.K537N.KLHL13:ENST00000371878.1:exon7:c.A1611T:p.K537N.KLHL13:ENST00000524145.1:exon8:c.A1716T:p.K588N.KLHL13:ENST00000541812.1:exon8:c.A1716T:p.K572N.KLHL13:ENST00000371876.1:exon8:c.A1611T:p.K537N.
X	12568573	G	A	TTCCACAGGTGAATGAGCG	exonic	XX	nonsense	DCAF12L1:ENST00000371126.1:exon1:c.C859T:p.H287Y.
X	129186538	G	A	CAGCTCAGAGAGGCTCAGCA	exonic	XX	nonsense	BCORL1:ENST00000459622.1:exon6:c.G321A:p.E1072K.BCORL1:ENST00000303743.5:exon6:c.G4414A:p.E1472K.
X	129333940	C	G	GCTGTGGCTGCTGCTCTCA	exonic	XX	nonsense	ELF4:ENST00000356977.7:exon8:c.G922C:p.E306Q.ELF4:ENST00000308167.5:exon8:c.G922C:p.E306Q.
X	130220344	G	A	AGCGCGTGTGAGTCCAGCC	exonic	XX	nonsense	ARHGAP36:ENST00000412432.2:exon10:c.G1230A:p.W410X.ARHGAP36:ENST00000370922.1:exon10:c.G1287A:p.W429X.ARHGAP36:ENST00000276211.5:exon10:c.G1323A:p.W441X.
X	130409948	C	T	TCACCCAGATATAAGGGGCA	exonic	XX	nonsense	IGSF1:ENST00000370603.3:exon15:c.C2968A:p.M960I.IGSF1:ENST00000370904.1:exon2:c.G2856A:p.M952I.IGSF1:ENST00000370910.1:exon14:c.G2856A:p.M952I.IGSF1:ENST00000361420.3:exon15:c.G2853A:p.M961I.
X	132181857	G	A	TTTGTGGAATAGTTTTGTT	exonic	XX	nonsense	USP26:ENST00000406273.1:exon2:c.C3927:p.S131L.USP26:ENST00000511190.1:exon6:c.C3927:p.S131L.USP26:ENST00000370832.1:exon1:c.C3927:p.S131L.
X	133378945	G	A	AAACAGTGCATAGCAGCAA	exonic	XX	nonsense	CCDC160:ENST00000370809.4:exon2:c.G115A:p.D39N.CCDC160:ENST00000517294.1:exon3:c.G115A:p.D39N.
X	135674131	G	A	ATTAATCCCAAGAGAAAT	exonic	XX	nonsense	BRS3:ENST00000370848.3:exon3:c.G797A:p.R206Q.
X	13668674	G	A	GTACTTTTACAAKCTCATGC	exonic	XX	nonsense	ATP11C:ENST00000316468.2:exon19:c.C2200T:p.P734S.ATP11C:ENST00000327569.3:exon19:c.C2200T:p.P734S.ATP11C:ENST00000359866.2:exon19:c.C2200T:p.P734S.ATP11C:ENST00000370557.1:exon19:c.C2191T:p.P731S.ATP11C:ENST00000370543.1:exon19:c.C2200T:p.P731S.
X	13603351	C	T	TGACAAATTCAGCTAATCA	exonic	XX	nonsense	CXorf66:ENST00000370540.1:exon3:c.G790A:p.E264K.
X	14099322	C	T	CTCCATTTTCAGAGTCTCT	exonic	XX	nonsense	MAGEC1:ENST00000285879.4:exon4:c.C2132T:p.P711L.
X	147743691	C	T	CCATGCTCTACCTCTGTT	exonic	XX	nonsense	AF2:ENST00000370458.1:exon3:c.C401T:p.P134L.AF2:ENST00000370457.5:exon3:c.C401T:p.P134L.AF2:ENST00000370460.2:exon3:c.C413T:p.P138L.AF2:ENST00000342251.3:exon3:c.C401T:p.P134L.
X	14803252	C	T	TGAGGCGACCTGTGGCAC	exonic	XX	nonsense	AF2:ENST00000286437.5:exon7:c.C403T:p.R155C.AF2:ENST00000370457.5:exon9:c.C1441T:p.R481C.AF2:ENST00000370460.2:exon10:c.C1540T:p.R514C.AF2:ENST00000342251.3:exon9:c.C1441T:p.R481C.
X	148638792	G	A	AGCAAGCAGGCTGCTGAC	exonic	XX	nonsense	MAML1:ENST00000432980.2:exon3:c.G872A:p.G291E.MAML1:ENST00000370405.1:exon2:c.G947A:p.G316E.MAML1:ENST00000292958.5:exon3:c.G947A:p.G316E.MAML1:ENST00000426613.2:exon2:c.G872A:p.G291E.
X	14902430	C	A	ACGATGGAATCACGGATGAG	exonic	XX	nonsense	MTMR1:ENST00000445323.2:exon15:c.C1840T:p.P814S.MTMR1:ENST0000041926.1:exon15:c.C1547:p.P912S.MTMR1:ENST00000370390.3:exon14:c.C1816T:p.P906S.MTMR1:ENST0000044228.1:exon14:c.C1816T:p.P906S.
X	151905076	G	C	GAGTATAGTATGGATGAG	exonic	XX	nonsense	MAGEA12:ENST00000357916.4:exon2:c.C225G:p.I75M.MAGEA12:ENST00000393900.3:exon3:c.C225G:p.I75M.MAGEA12:ENST00000393869.3:exon3:c.C225G:p.I75M.
X	15312458	G	A	CCAGCAGGAGGACCTCAGG	exonic	XX	nonsense	L1CAM:ENST00000538883.1:exon25:c.C3343T:p.P11155L.L1CAM:ENST00000543994.1:exon25:c.C3343T:p.P11155L.L1CAM:ENST00000370055.1:exon25:c.C3322T:p.P11085.L1CAM:ENST00000361981.3:exon24:c.C3322T:p.P11085.L1CAM:ENST00000370057.3:exon25:c.C3337T:p.I937Y.
X	15504137	G	C	CTCCAGTATATGAGACAA	exonic	XX	nonsense	SPRY3:ENST00000302805.2:exon2:c.G904C:p.D209H.

=large_intestine

c.13262C>AAChange-p.P4421t.tissue=lung

135G>AAChange-p.E379K.tissue=large_intestine&NEK10_ENST0000429845;basechange=c.1135G>AAChange-p.E379K.tissue=large_intestine

je-c:5074G>A.AChange+p.D1692N:issue=lung

>T.AChange+p.V757F:issue=kidney

je-c:1282C>A.AChange+p.P428T:issue=lung

change>p.V181SF.tissue=lung

basechange=c.1638C>TAAchange=p.S613L.tissue=urinary_tract

urinary_tract

U>C.AAchange=p.N400H.tissue=large_intestine&DCLK1_ENST00000379893.basechange=c.277A>C.AAchange=p.N93H.tissue=large_intestine

rg=c.3704C>TAAchange-p.S1235F.tissue=urinary_tract.large_intestine

>TAAchange-p.R309C.tissue=large_intestine

Chromosome	Position (hg)	Reference or db	Mutant Allele/Sequence context	ANNOVAR RefSeq	Mutation type	ANNOVAR output (alternative nomenclature)	Cytoband	COSMIC
1	116343 G	A	CCTCTGCTGTAAAGCGGGGACT	SNV	SNV	SNV		
1	116841 G	T	CTGGGGCCCTGCTGGGGCCG	SNV	SNV	SNV		
1	123801F10	A	CCGACTCTCCGCTCTGCTGGCAG	SNV	SNV	SNV		
1	1410736T	A	TCCGACTTCTGCCCAACAGT	SNV	SNV	SNV		
1	162550C	T	AGCTGTAGTACTACTCTCCCT	SNV	SNV	SNV		
1	162561F1	G	GAAGAAGTCGCCCTTACTCT	SNV	SNV	SNV		
1	1626136C	T	GGCGCCACCATTACAGCTT	SNV	SNV	SNV		
1	1944780G	T	CAGGAGAGCAATCCAGCCAGC	SNV	SNV	SNV		
1	232620F1	C	CCAGCGGCTTTATATGCTGGA	SNV	SNV	SNV		
1	2381057G	C	TGACGGCCAGCAATCCGGCGGA	SNV	SNV	SNV		
1	241847A	G	GAGCAGAGCAATCCAGCCAGC	SNV	SNV	SNV		
1	256837G	A	CCGCTCCAGCAGCAATCCAGCA	SNV	SNV	SNV		
1	2828876G	A	TCTGTCTGCAAGGAAGCGCA	SNV	SNV	SNV		
1	2947586G	T	GGTCTTCCGCAATCTCATCTG	SNV	SNV	SNV		
1	332075F10	T	TCGAGAGTTTATTGATCTCTC	SNV	SNV	SNV		
1	3656370G	A	GGGGGTCCCGGGCCGGCGGGA	SNV	SNV	SNV		
1	4326185C	T	GCTGTCTCTGTATGTCTCAGG	SNV	SNV	SNV		
1	4365102A	C	GCAGAGAAATAGGCTAAGCC	SNV	SNV	SNV		
1	4365127A	C	CAAGTTCACATCAGCCCTCAT	SNV	SNV	SNV		
1	4368885G	T	GGTCTCTCTCTGGTGGGGCT	SNV	SNV	SNV		
1	4406965G	A	GGGAGGACCGGGCCATCAT	SNV	SNV	SNV		
1	526347C	T	CGTTTACAGAGGGCATCTCA	SNV	SNV	SNV		
1	5323665G	T	AAGCTTCTCTCAAGCCCAAG	SNV	SNV	SNV		
1	5447783A	T	GACAGGGCTCTAGTAAAGACT	SNV	SNV	SNV		
1	6049262A	C	AATCTCAACAGCACTCTGT	SNV	SNV	SNV		
1	7192007F10	T	TGCGTCTTATCTAGTACGCT	SNV	SNV	SNV		
1	9364571G	T	GGCAGATCTTGGCCCATCC	SNV	SNV	SNV		
1	11066997G	A	GGCTCAGCCCTACTGTAGGTCA	SNV	SNV	SNV		
1	12030182A	T	AAGCTCTCCCCTTCATATTG	SNV	SNV	SNV		
1	15403092G	C	TCGAACTGAACTGGATGGCTG	SNV	SNV	SNV		
1	15523320G	T	TGGACTCTTGGCCGGGCTT	SNV	SNV	SNV		
1	15664171T	G	TTTGAAGGAGTTCTCAATCTG	SNV	SNV	SNV		
1	15664171T	T	GGGCGGGCTCCAGCAGCCAGC	SNV	SNV	SNV		
1	15667178A	T	CGGCTGGGCTGCTTGGATCT	SNV	SNV	SNV		
1	15773833G	C	TCTTACGACAGATGTGATGC	SNV	SNV	SNV		
1	16177186A	A	CCCTTCCAGTATAGCCAGCAG	SNV	SNV	SNV		
1	16189249G	C	AGCTTCAGAGATCCAGCGCCA	SNV	SNV	SNV		
1	16262586C	A	TTTGAAGCAGCATATGATCTCA	SNV	SNV	SNV		
1	16951087C	T	TCTCACTGATCTTGGAGAAG	SNV	SNV	SNV		
1	17361681G	C	CTCAATAAATCTTCAATCTG	SNV	SNV	SNV		
1	17680392F10	A	GGCTCTGCTCTCAACTCTCTCC	SNV	SNV	SNV		
1	17902740G	C	GGAAGCTCACTAAACTACTCG	SNV	SNV	SNV		
1	18044140C	T	AGTTTATTAATCTGATCTGAT	SNV	SNV	SNV		
1	1826276F10	A	TGAGAGCTCTCACTCCAGCAC	SNV	SNV	SNV		
1	20103467T	A	CTCTCAGCCCTGGCCCTTGT	SNV	SNV	SNV		
1	20195996C	T	GAGAGAGGATCCAGCAAAAT	SNV	SNV	SNV		
1	20117523G	A	ATGGCTCTCTCAAGCAGTCT	SNV	SNV	SNV		
1	20182540A	C	ATATCTCTCTCATGCAATC	SNV	SNV	SNV		
1	20271981T	T	CTTACAGCCCTCCAGCAATAT	SNV	SNV	SNV		
1	20464852C	T	AGACAGCTCTACCTCTGGCT	SNV	SNV	SNV		
1	20512917F10	T	GAGGGCCAGCAAGGATGCCC	SNV	SNV	SNV		
1	20539436G	T	TTGAGAGCCCTCAGCACTGGC	SNV	SNV	SNV		
1	20987834A	C	GCAAGCAGGATTCAGCAGCC	SNV	SNV	SNV		
1	22262651C	G	CTTTGACTAATGCATTAACAC	SNV	SNV	SNV		
1	22664786C	T	CTGTTGGGATGATCCAGCCGAA	SNV	SNV	SNV		
1	23007181C	G	GATCAGGTTACATGCAGAGC	SNV	SNV	SNV		
1	2329790G	A	AGCATCTCACAGCATATGCT	SNV	SNV	SNV		
1	23294238C	splicing	AGCATCTCTCTAGAAATGGA	SNV	SNV	SNV		
1	24025595G	C	CTCCGGCGAATGACTCTGGA	SNV	SNV	SNV		
1	24795846C	T	TTTGAAGCTTCCAGCCGAGCA	SNV	SNV	SNV		
1	271346T	G	AGAACAGCCGAACTGAGAGCA	SNV	SNV	SNV		
1	282958G	T	TCTCTGGGCTGGCTCTCAC	SNV	SNV	SNV		
1	2841432A	A	TTTCTGAGAAAGAGAGAGACA	SNV	SNV	SNV		
1	2732422G	T	GTTCCCTGCAAGTCTGGCT	SNV	SNV	SNV		
1	2772400F10	G	TGAGCGTCCCTCTGGCGAGG	SNV	SNV	SNV		
1	37193648G	T	TTGAGAGCCCTCAGCACTGGC	SNV	SNV	SNV		
1	44428113C	C	ATACAGCCTGTAAATGTTGCA	SNV	SNV	SNV		
1	45260374C	A	GTCAGCTCAAGCTCTGCGTGA	SNV	SNV	SNV		
1	5517778F10	A	CTGCTGCTCTACACAGCTTT	SNV	SNV	SNV		
1	5617867F10	C	GGAAGCAGGCGGATGACT	SNV	SNV	SNV		
1	5136232T	C	AAGATCTGCAATCTCTAAC	SNV	SNV	SNV		
1	5146196C	T	TGAGAGACGGAATGCAAGCT	SNV	SNV	SNV		
1	52497318C	T	CTGCTCTCAATGGCTTTT	SNV	SNV	SNV		
1	5366442T	A	TTTCTGAGAAATATCTTCT	SNV	SNV	SNV		
1	54954293G	T	TTTCTGAGCAAACTCTCCAG	SNV	SNV	SNV		
1	56591608C	T	TCTGAGGAAATCCATCTCT	SNV	SNV	SNV		
1	56581011G	T	GGAAGCAACTATCCAGCTGG	SNV	SNV	SNV		
1	57021189G	T	CGGGCGGCGAGCGCCGGGCT	SNV	SNV	SNV		
1	58081026C	A	GTTTTTCTGCTCTCTTCTC	SNV	SNV	SNV		
1	59132923C	T	TAGCAGAAATAGGAGCAGG	SNV	SNV	SNV		
1	20326297G	A	AGCCAAAGAGAGAGAAGG	SNV	SNV	SNV		
1	2115590F10	T	GGTCTCTTCTTTGGAATGG	SNV	SNV	SNV		
1	21193302C	T	AGAGCTCAAGCTGATGAGCT	SNV	SNV	SNV		
1	21949283G	A	TGGATGATAGTGGAGCACT	SNV	SNV	SNV		
1	23469494C	T	AGCTCCATGAGCTGGGACT	SNV	SNV	SNV		
1	24292311T	T	CTGTCTCTGGTGGCCAGCA	SNV	SNV	SNV		
1	3915778A	A	AGTTAAAGATCCAGCGGACT	SNV	SNV	SNV		
1	4263869C	B	TTCTGTGAGCTCTCATCCAG	SNV	SNV	SNV		
1	4472584G	A	GAGCTGTGTTGAAGGTGGG	SNV	SNV	SNV		
1	3703369A	G	TATCGCTCCCTCTTGTGAAA	SNV	SNV	SNV		
1	38729648A	T	GATCGCTGGATCTGGAGGCC	SNV	SNV	SNV		
1	4278470G	C	GATAGAGACTCTGAAGCTGT	SNV	SNV	SNV		
1	4428494A	T	TCCACATGAAAATAAATAA	SNV	SNV	SNV		
1	4566552G	T	CCCGCCGCTTGGATGCTTT	SNV	SNV	SNV		
1	4566529A	T	TCCAGGAGGCTCTATTTCTC	SNV	SNV	SNV		
1	4744651C	G	ATCTGATCTAGCACCACTG	SNV	SNV	SNV		
1	4871631G	T	CCACCCCTCTTTCAGGAGCA	SNV	SNV	SNV		
1	4965320F10	C	TGTACCTGGGCTGTAGAGAG	SNV	SNV	SNV		
1	4989460G	B	GTTCTCTCTGCGCAGCAAGT	SNV	SNV	SNV		
1	4975648G	C	AGCTCCGACTCCAGCGGGCA	SNV	SNV	SNV		
1	4982378G	A	CTCTTACGGCTCTGGTCTGCG	SNV	SNV	SNV		
1	5164096C	A	ACTGCGCAAAAGGTTCTCT	SNV	SNV	SNV		
1	5384274C	A	GTTCCGCTGTCTCATCTCT	SNV	SNV	SNV		
1	5724571G	T	TAAGAAAGCAATCCAGAGC	SNV	SNV	SNV		
1	8546484C	C	TGTATATTTAATGCACTCT	SNV	SNV	SNV		
1	11272469A	G	GTACATGTTGAGTCTCCAC	SNV	SNV	SNV		
1	1193191F10	T	AGATCAACTCAACTGAGCT	SNV	SNV	SNV		
1	11928990C	T	AGCAGAGGCTGGTCAAGGTT	SNV	SNV	SNV		
1	12148178F10	T	GATGATGATCTAGTAAACT	SNV	SNV	SNV		
1	1357161A	A	CAAAATATGAGGCAACTGCTA	SNV	SNV	SNV		
1	14030721T	C	AAAGAAATAATCAACTACAC	SNV	SNV	SNV		
1	14116322A	A	TCCATCACTAAGGAAAGGAA	SNV	SNV	SNV		
1	14116354G	A	TCCATCACTAAGGAAAGGAA	SNV	SNV	SNV		
1	14215379F10	T	TGCTCAACTTATCAACTTCA	SNV	SNV	SNV		
1	14630754C	T	AGCATGATTAATACTGACAC	SNV	SNV	SNV		
1	15106612T	A	AATGCCAGCAAGCAGCAGC	SNV	SNV	SNV		
1	15106624G	A	AAATGGCTCTGGTCTCTG	SNV	SNV	SNV		
1	15623863G	T	AGCTTCTCACCTCCAGCAGC	SNV	SNV	SNV		
1	15653976C	T	AGCAGCAGCACTTGTCTCC	SNV	SNV	SNV		
1	16470168T	A	TTCAGCAGGAGTCTCTCTCT	SNV	SNV	SNV		
1	16480666C	T	TGAAGAACCAACTGCTCTCT	SNV	SNV	SNV		
1	17283542G	T	TTTCTGTCTTGTATTTCT	SNV	SNV	SNV		
1	17324279G	T	CCCATTTACTCTTATGATTT	SNV	SNV	SNV		
1	18644335A	T	GATATATCTGCTTATAGAGC	SNV	SNV	SNV		
1	19418658A	C	TACTTATTATTAATGCTTAT	SNV	SNV	SNV		
1	2267022C	T	CCAGCCAGCGTCCCGGGCT	SNV	SNV	SNV		
1	303919T	C	CAGTCTCCGGTGGTGAAGGG	SNV	SNV	SNV		
1	5570268A	A	GCCAGCAGCTAGCTGTGAG	SNV	SNV	SNV		
1	563835C	T	CTATTCCTCTCTATATCCG	SNV	SNV	SNV		
1	6302919G	A	CTTCTTCACTCAGCAACTC	SNV	SNV	SNV		
1	7275559G	A	AGGCCAGCACTAGGCCGCT	SNV	SNV	SNV		
1	1301331G	A	CAGCGCAGCCCTTCTTGGC	SNV	SNV	SNV		
1	37488730G	T	TTTCTATTTAGGTAGGTTG	SNV	SNV	SNV		
1	3844856C	T	GGGACTGCGGGAAGCCCGC	SNV	SNV	SNV		
1	4162265G	T	GAGCCCTCTATGCTGGGTT	SNV	SNV	SNV		
1	4248787C	T	AAGGATAGTAAAGTCAAGCT	SNV	SNV	SNV		
1	42571221G	C	TACAGGCACTGCTTCAAGAA	SNV	SNV	SNV		
1	4784754T	A	TGCTTATCACTGAGCAGGAG	SNV	SNV	SNV		
1	4788793G	C	TCCATGACAGAGGAGGACAC	SNV	SNV	SNV		
1	71495242G	A	GTCGAGGCTTCAACTCTCTT	SNV	SNV	SNV		
1	8563962A	T	CTCACTGGTCTAGGCGCT	SNV	SNV	SNV		
1	96104115C	G	ACAGATTGTAAACAGAGCTC	SNV	SNV	SNV		
1	9676904G	T	TATCTCTCTGCTAAAGGTC	SNV	SNV	SNV		
1	1005704							

196?issue=ovary.breast.stomach.haematopoietic_and_lymphoid_tissue.urinary_tract.kidney.pancreas.liver.oesophagus.skin.lung.upper_aerodigestive_tract.large_intestine.central_nervous_system.biliary_tract&TP53_ENST00000269305.basecharge=c.586C>T.AAchange+p.R119?issue=breast.stomach.haematopoietic_and_lymphoid_tissue.oesophagus.lung.upper_aerodigestive_tract.large_intestine.central_nervous_system.biliary_tract&TP53_ENST00000455263.basecharge=c.586C>T.AAchange+p.R119?issue=stomach.lung.large_intestine&TP53_ENST00000413465.basecharge=c.586C>T.AAchange+p.R119?issue=breast.haematopoietic_and_lymphoid_tissue.stomach.oesophagus.lung.upper_aerodigestive_tract.large_intestine

Supplemental Table 4

cytoband	1q21.3	8q24.3	7p22.1	21q22.3
q value	0,0006579	0,0006579	0,091895	0,091895
residual q value	0,0006579	0,0006579	0,091895	0,091895
wide peak boundaries	chr1:152265562-152329851	chr8:144980719-145001933	chr7:6744791-6864178	chr21:45953569-46082924
genes in wide peak	FLG FLG2	PLEC	ZNF12 CCZ1B RSPH10B PMS2CL RSPH10B2	TSPEAR KRTAP10-10 KRTAP10-4 KRTAP10-6 KRTAP10-7 KRTAP10-9 KRTAP10-1 KRTAP10-11 KRTAP10-2 KRTAP10-5 KRTAP10-8 KRTAP10-3 KRTAP12-3 KRTAP12-4

cytoband	5q22.2	2q32.2	2q36.3	9q22.33	15q21.1	1p21.1	6q13	10q23.31	12q24.33	3p24.1	4q13.3	13q13.1	22q11.21	10p12.33	1p22.3
q value	1,6726E-08	1,6726E-08	0,000072259	0,00081909	0,0021785	0,0044551	0,0062561	0,0062561	0,013069	0,013306	0,029455	0,041181	0,060988	0,085556	0,099783
residual															
q value	1,6726E-08	1,7698E-08	0,00042243	0,00081909	0,0021785	0,0044551	0,0062561	0,0062561	0,013069	0,013306	0,029354	0,041181	0,060988	0,085556	0,10347
wide															
peak															
boundaries	chr5:1117476 67-112200958	chr2:1898768 82-190014430	chr2:2278531 64-228192407	chr9:1018295 55-101983418	chr15:486301 95-49034995	chr1:1024540 27-104074629	chr6:705027 82-71133026	chr10:896112 76-90034788	chr12:1334576 37-133851895	chr3:300433 44-31205400	chr4:706207 36-71204641	chr13:328757 88-32993270	chr22:213807 10-21922059	chr10:177546 11-18292099	chr1:861667 05-86820520
genes in															
wide															
peak	APC FLJ11235	hsa-mir-3129 MIR3129	COL4A3 COL4A4	TGFBR1	FBN1	COL11A1	COL9A1 COL19A1	PTEN KLLN	ZNF10 ZNF26 ZNF84 ZNF140 ZNF268 LOC647589 ZNF605	hsa-mir-466 TGFBR2 GADL1	CSN1S1 CSN2 CSN3 HTN1 HTN3 STATH SULT1E1 ODAM CABS1 FDCSP CSN1S2AP C4orf40 CSN1S2BP	BRCA2 ZAR1L	hsa-mir-649 SLC7A4 HIC2 POM121L8P RIMBP3C PI4KAP2 LOC400891 BCRP2 P2RX6P RIMBP3B TMEM191C	hsa-mir-511-2 hsa-mir-511-1 MRC1 MIR511-1 MIR511-2 TMEM236	COL24A1

Supplemental Table 5

	AC1	AC2	AC3	AC4	AC6	AC7	AC8	AC11	AC12	AC13	AC14	AC17	AC19	AC21	AC23	AC25	AC27	AC29	AC30
C000-6KNGDL						60		25					15						
C000-X88E7Z		17				39		17		26									
C000-RCWM1J				7		52		41											
C000-EXKBCS				15		49		29							7				
C000-HZGGV2		5		5		49		32		7		2							
C000-ESMKCH				15		41		44											
C000-S5ZHWR		7				40		31		12		10							
C000-2SXE39			30	30						4			11	9		12	3		
C000-ZZP8UU	5	15	66										11				4		
C000-CFLP5N	4	11	44			6		16					8						10
C000-JMUDMJ		16				28		22		20				13					
C000-648QQH	4	11	18		6	9	26		27										
C000-W91YB6	43	10						16		31									
C000-AX1WLG						61				9	8							22	

Relative signature contributions (% of SNVs attributed to the signature)