SUPPLEMENTAL MATERIAL

Kiel et al., http://www.jem.org/cgi/content/full/jem.20120910/DC1

Table S1. **Genes involved in structural alterations**

Genome	LeftChr	LeftPosition	LeftStrand	Left gene	RightChr	RightPosition	RightStrand	Right gene	Interchromosomal	StrandConsistent	Distance	Displayed
A01	chr1	10,543,646	+	PEX14	chr1	10,546,089	+	PEX14	N	Y	2,443	
A01	chr1	78,833,897	+	MGC27382	chr1	78,835,838	+		N	Y	1,941	
A01	chr1	246,368,887	+	SMYD3	chr1	246,388,776	+	SMYD3	N	Y	19,889	
A01	chr2	41,913,661	+		chr6	13,191,446	-	PHACTR1	Y	N		yes
A01	chr2	51,748,064	-		chr6	117,811,873	-	DCBLD1	Y	Y		yes
A01	chr2	51,748,196	+		chr6	117,811,877	+	DCBLD1	Y	Y		yes
A01	chr2	55,634,190	+	CCDC88A	chr2	55,636,191	+	CCDC88A	N	Y	2,001	
A01	chr2	77,657,786	+	LRRTM4	chr2	77,659,139	+	LRRTM4	N	Y	1,353	
A01	chr2	144,011,246	+	ARHGAP15	chr6	154,152,886	-		Y	N		yes
A01	chr2	175,507,361	+	WIPF1	chr2	175,508,424	+	WIPF1	N	Y	1,063	
A01	chr3	9,696,986	+	MTMR14	chr3	9,697,814	+	MTMR14	N	Y	828	
A01	chr3	30,865,778	+	GADL1	chr3	30,866,269	+	GADL1	N	Y	491	
A01	chr3	120,161,644	+	FSTL1	chr3	120,164,951	+	FSTL1	N	Y	3,307	
A01	chr3	123,135,519	+	ADCY5	chr3	123,136,265	+	ADCY5	N	Y	746	
A01	chr3	152,879,998	-		chrX	76,982,473	-	ATRX	Y	Y		yes
A01	chr3	172,715,821	-	SPATA16	chr3	173,133,709	+	NLGN1	N	N	417,888	
A01	chr4	10,544,912	+	CLNK	chr4	10,552,747	+	CLNK	N	Y	7,835	
A01	chr4	17,982,341	+	LCORL	chr4	17,983,912	+	LCORL	N	Y	1,571	
A01	chr4	83,636,008	+	SCD5	chr4	83,641,270	+	SCD5	N	Y	5,262	
A01	chr4	113,564,756	-	LARP7	chr4	113,578,883	-		N	Y	14,127	
A01	chr4	144,298,455	+	GAB1	chr4	144,299,530	+	GAB1	N	Y	1,075	
A01	chr4	144,307,924	+	GAB1	chr4	144,308,523	+	GAB1	N	Y	599	
A01	chr5	16,869,967	+	MYO10	chr5	16,870,995	+	MYO10	N	Y	1,028	
A01	chr5	80,354,955	+	RASGRF2	chr5	80,355,664	+	RASGRF2	N	Y	709	
A01	chr5		+	FGF1	chr5		+	FGF1	N	Y		

		141,999,406				141,999,982					576
A01	chr6	80,993,706	+	BCKDHB	chr6	80,999,802	+	BCKDHB	N	Y	6,096
A01	chr7	55,913,756	+	40800	chr7	55,917,452	+	40800	N	Y	3,696
A01	chr7	100,341,310	+	ZAN	chr7	100,343,319	+	ZAN	N	Y	2,009
A01	chr7	117,455,132	+	CTTNBP2	chr7	117,459,638	+	CTTNBP2	N	Y	4,506
A01	chr7	134,919,336	+	STRA8	chr7	134,920,501	+	STRA8	N	Y	1,165
A01	chr7	140,231,450	+	DENND2A	chr7	140,231,914	+	DENND2A	N	Y	464
A01	chr7	157,671,001	+	PTPRN2	chr7	157,671,947	+	PTPRN2	N	Y	946
A01	chr8	124,958,894	+	FER1L6	chr8	124,961,978	+	FER1L6	N	Y	3,084
A01	chr9	642,168	+	KANK1	chr9	648,239	+	KANK1	N	Y	6,071
A01	chr9	119,513,452	-	ASTN2	chr9	119,515,996	-	ASTN2	N	Y	2,544
A01	chr9	126,616,998	+	DENND1A	chr9	126,617,683	+	DENND1A	N	Y	685
A01	chr10	68,169,701	+	CTNNA3	chr10	68,217,728	+	CTNNA3	N	Y	48,027
A01	chr10	76,888,531	+	SAMD8	chr10	76,895,652	+	SAMD8	N	Y	7,121
A01	chr11	19,079,343	+	MRGPRX2	chr11	19,080,395	+	MRGPRX2	N	Y	1,052
A01	chr11	19,079,350	-	MRGPRX2	chr11	19,080,724	-	MRGPRX2	N	Y	1,374
A01	chr11	65,933,634	+	PACS1	chr11	65,939,063	+	PACS1	N	Y	5,429
A01	chr11	66,657,990	+	PC	chr11	66,658,904	+	PC	N	Y	914
A01	chr11	71,660,390	+	RNF121	chr11	71,660,973	+	RNF121	N	Y	583
A01	chr12	44,691,832	+	TMEM117	chr12	44,692,914	+	TMEM117	N	Y	1,082
A01	chr12	53,595,998	+	ITGB7	chr12	53,596,600	+	ITGB7	N	Y	602
A01	chr12	86,695,694	+	MGAT4C	chr12	86,703,066	+	MGAT4C	N	Y	7,372
A01	chr12	129,787,758	+	TMEM132D	chr12	129,788,178	+	TMEM132D	N	Y	420
A01	chr13	26,024,956	+	ATP8A2	chr13	26,026,278	+	ATP8A2	N	Y	1,322
A01	chr13	93,363,496	+	GPC5	chr13	93,364,985	+	GPC5	N	Y	1,489
A01	chr14	33,603,568	+	NPAS3	chr14	33,632,183	+	NPAS3	N	Y	28,615
A01	chr14	79,159,147	+	NRXN3	chr14	79,165,647	+	NRXN3	N	Y	6,500
A01	chr16	131,680	+	MPG	chr16	132,250	+	MPG	N	Y	570
A01	chr16	81,407,483	-	GAN	chr16	81,408,094	-	GAN	N	Y	611
A01	chr17	2,398,975	+	METTL16	chr17	2,402,799	+	METTL16	N	Y	3,824
A01	chr17	33,681,081	+	SLFN11	chr17	33,689,757	+	SLFN11	N	Y	8,676

A01	chr17	33,687,392	+	SLFN11	chr17	33,689,759	+	SLFN11	N	Y	2,367	
A01	chr17	33,690,847	+	SLFN11	chr17	33,700,494	+	SLFN11	N	Y	9,647	
A01	chr17	73,052,506	-	KCTD2	chr17	73,054,209	-	KCTD2	N	Y	1,703	
A01	chr19	17,794,376	+	UNC13A	chr19	17,794,814	+	UNC13A	N	Y	438	
A01	chr19	23,856,633	+	ZNF675	chr19	23,866,242	+	ZNF675	N	Y	9,609	
A01	chr19	53,477,443	+	ZNF702P	chr19	53,477,962	+	ZNF702P	N	Y	519	
A01	chr22	49,074,729	+	FAM19A5	chr22	49,075,600	+	FAM19A5	N	Y	871	
A01	chrX	2,351,840	-	DHRSX	chrX	2,355,063	-	DHRSX	N	Y	3,223	
A01	chrX	135,331,945	+	MAP7D3	chrX	135,332,550	+	MAP7D3	N	Y	605	
B01	chr2	77,686,969	-	LRRTM4	chr2	77,693,012	-	LRRTM4	N	Y	6,043	
B01	chr2	77,687,039	+	LRRTM4	chr2	77,693,083	+	LRRTM4	N	Y	6,044	
B01	chr2	143,926,139	+	ARHGAP15	chr2	143,927,714	+	ARHGAP15	N	Y	1,575	
B01	chr4	71,802,258	+	MOBKL1A	chr4	71,804,751	+	MOBKL1A	N	Y	2,493	
B01	chr7	138,363,473	-	SVOPL	chr7	138,365,051	-		N	Y	1,578	
B01	chr7	157,769,125	+	PTPRN2	chr7	157,772,013	+	PTPRN2	N	Y	2,888	
B01	chr9	21,842,086	+	MTAP	chr9	21,854,133	+	MTAP	N	Y	12,047	
B01	chr9	22,252,932	-		chr13	67,763,729	+	PCDH9	Y	N		yes
B01	chr10	103,445,737	+	FBXW4	chr10	103,446,338	+	FBXW4	N	Y	601	
B01	chr11	84,563,723	-	DLG2	chr11	84,565,261	-	DLG2	N	Y	1,538	
B01	chr11	84,563,800	+	DLG2	chr11	84,565,277	+	DLG2	N	Y	1,477	
B01	chr11	94,080,417	-		chrX	73,718,032	-	SLC16A2	Y	Y		yes
B01	chr12	53,595,998	+	ITGB7	chr12	53,596,600	+	ITGB7	N	Y	602	
B01	chr12	70,679,895	+	CNOT2	chr12	70,680,974	-	CNOT2	N	N	1,079	
B01	chr12	70,680,612	-	CNOT2	chr12	70,682,065	+	CNOT2	N	N	1,453	
B01	chr12	104,730,691	+	TXNRD1	chr12	104,732,448	+	TXNRD1	N	Y	1,757	
B01	chr13	21,730,905	+	SKA3	chr13	21,731,227	+	SKA3	N	Y	322	
B01	chr14	79,159,149	+	NRXN3	chr14	79,165,649	+	NRXN3	N	Y	6,500	
B01	chr17	9,749,059	+	GLP2R	chr17	9,749,587	+	GLP2R	N	Y	528	
B01	chr17	33,681,081	+	SLFN11	chr17	33,689,757	+	SLFN11	N	Y	8,676	
B01	chr17	33,690,846	+	SLFN11	chr17	33,700,493	+	SLFN11	N	Y	9,647	
B01	chr18		+	PIK3C3	chr18		+	PIK3C3	N	Y		

		39,626,845				39,627,703					858	
B01	chr19	17,359,252	+		chr19	17,361,016	+	USHBP1	N	Y	1,764	
B01	chr19	37,922,139	+	ZNF569	chr19	38,018,371	+	ZNF793	N	Y	96,232	
B01	chr21	22,852,029	+	NCAM2	chr21	22,852,660	+	NCAM2	N	Y	631	
B01	chr21	36,203,887	+	RUNX1	chr21	36,204,890	+	RUNX1	N	Y	1,003	
B01	chr22	17,257,507	+		chr22	17,273,377	+	XKR3	N	Y	15,870	
B01	chr22	34,308,986	-	LARGE	chr22	34,311,623	+	LARGE	N	N	2,637	
B01	chr22	34,309,375	+	LARGE	chr22	34,309,889	-	LARGE	N	N	514	
C01	chr1	53,499,006	-	SCP2	chr1	53,499,667	-	SCP2	N	Y	661	
C01	chr1	159,020,405	+	IFI16	chr1	159,021,049	+	IFI16	N	Y	644	
C01	chr1	235,351,893	+	ARID4B	chr1	235,355,320	+	ARID4B	N	Y	3,427	
C01	chr2	153,491,852	+	FMNL2	chr2	153,493,458	-	FMNL2	N	N	1,606	
C01	chr2	153,492,542	-	FMNL2	chr2	153,495,278	+	FMNL2	N	N	2,736	
C01	chr2	187,015,782	+		chr7	99,015,637	-	BUD31	Y	N		yes
C01	chr2	187,019,774	-		chr7	99,015,451	+	BUD31	Y	N		yes
C01	chr4	152,732,426	-		chr5	121,670,726	-	SNCAIP	Y	Y		yes
C01	chr4	152,732,633	+		chr5	121,670,722	+	SNCAIP	Y	Y		yes
C01	chr5	63,885,142	+	RGS7BP	chr5	63,886,316	+	RGS7BP	N	Y	1,174	
C01	chr5	129,480,450	+	CHSY3	chr5	129,481,193	+	CHSY3	N	Y	743	
C01	chr6	102,427,787	+	GRIK2	chr6	102,428,220	+	GRIK2	N	Y	433	
C01	chr7	4,252,922	+	SDK1	chr7	4,253,863	+	SDK1	N	Y	941	
C01	chr7	110,121,351	-		chr7	110,384,334	-	IMMP2L	N	Y	262,983	
C01	chr7	120,494,993	+	TSPAN12	chr7	120,496,061	+	TSPAN12	N	Y	1,068	
C01	chr9	15,231,269	+	TTC39B	chr9	15,371,978	+		N	Y	140,709	
C01	chr9	131,556,898	+	TBC1D13	chr9	131,557,883	+	TBC1D13	N	Y	985	
C01	chr10	56,445,983	-	PCDH15	chr10	56,469,598	-	PCDH15	N	Y	23,615	
C01	chr10	88,050,480	+	GRID1	chr10	90,938,333	-		N	N	2,887,853	yes
C01	chr10	88,716,882	+	MMRN2	chr10	89,537,630	-	ATAD1	N	N	820,748	yes
C01	chr10	90,125,761	+	RNLS	chr10	90,690,776	-		N	N	565,015	yes
C01	chr10	117,655,951	-	ATRNL1	chr10	117,660,063	-	ATRNL1	N	Y	4,112	
C01	chr11	34,172,148	-		chr11	34,174,106	-	ABTB2	N	Y	1,958	

C01	chr11	121,962,273	+	MIR100HG	chr11	122,722,674	+	CRTAM	N	Y	760,401	yes
C01	chr12	53,595,998	+	ITGB7	chr12	53,596,600	+	ITGB7	N	Y	602	
C01	chr17	18,234,030	-	SHMT1	chr17	18,234,369	-	SHMT1	N	Y	339	
C01	chr17	33,681,092	+	SLFN11	chr17	33,689,757	+	SLFN11	N	Y	8,665	
C01	chr17	33,687,392	+	SLFN11	chr17	33,689,759	+	SLFN11	N	Y	2,367	
C01	chr17	33,690,846	+	SLFN11	chr17	33,700,493	+	SLFN11	N	Y	9,647	
C01	chr17	43,364,665	+	MAP3K14	chr17	43,372,175	+	MAP3K14	N	Y	7,510	
C01	chr18	77,869,498	-	ADNP2	chr18	77,929,015	-	PARD6G	N	Y	59,517	
C01	chr22	37,415,327	+	TST	chr22	37,420,695	+	MPST	N	Y	5,368	
D01	chr1	172,282,604	+	DNM3	chr1	172,291,753	+	DNM3	N	Y	9,149	
D01	chr2	173,302,495	+	ITGA6	chr2	173,304,472	+	ITGA6	N	Y	1,977	
D01	chr3	100,334,873	-	GPR128	chr3	100,446,152	-	TFG	N	Y	111,279	
D01	chr3	123,135,519	+	ADCY5	chr3	123,136,265	+	ADCY5	N	Y	746	
D01	chr5	129,020,376	+	ADAMTS19	chr5	129,024,195	+	ADAMTS19	N	Y	3,819	
D01	chr5	149,230,181	-	PPARGC1B	chr5	149,270,199	-	PDE6A	N	Y	40,018	
D01	chr7	18,826,300	+	HDAC9	chr7	18,826,467	+	HDAC9	N	Y	167	
D01	chr8	57,048,719	-		chr8	57,098,539	-	PLAG1	N	Y	49,820	
D01	chr9	113,667,553	+	LPAR1	chr9	113,669,264	+	LPAR1	N	Y	1,711	
D01	chr10	105,128,324	+	TAF5	chr10	105,133,114	+	TAF5	N	Y	4,790	
D01	chr14	47,672,012	+	MDGA2	chr14	47,678,230	+	MDGA2	N	Y	6,218	
D01	chr15	85,380,985	-	ALPK3	chr15	85,381,400	+	ALPK3	N	N	415	
D01	chr15	85,381,131	+	ALPK3	chr15	85,381,398	-	ALPK3	N	N	267	
D01	chr16	83,196,147	+	CDH13	chr16	83,209,726	+	CDH13	N	Y	13,579	
D01	chr17	44,887,353	+	WNT3	chr17	44,887,685	+	WNT3	N	Y	332	
D01	chr21	43,703,913	+	ABCG1	chr21	43,704,397	+	ABCG1	N	Y	484	
D01	chr22	46,924,695	+	CELSR1	chr22	46,925,569	+	CELSR1	N	Y	874	
E01	chr1	162,378,221	+	SH2D1B	chr1	162,378,877	+	SH2D1B	N	Y	656	
E01	chr2	32,201,502	+	MEMO1	chr2	32,203,192	+	MEMO1	N	Y	1,690	
E01	chr2	46,128,512	+	PRKCE	chr2	46,132,406	+	PRKCE	N	Y	3,894	
E01	chr2	206,246,836	+	PARD3B	chr2	206,255,783	+	PARD3B	N	Y	8,947	
E01	chr4		-		chr6		+	BACH2	Y	N		yes

		6,635,439				90,979,006						
E01	chr4	6,635,748	+		chr6	90,978,997	-	BACH2	Y	N		yes
E01	chr4	128,964,985	-		chr17	49,977,259	-	CA10	Y	Y		yes
E01	chr5	9,267,219	+	SEMA5A	chr5	9,275,423	+	SEMA5A	N	Y	8,204	
E01	chr6	90,979,006	-	BACH2	chr17	70,860,680	-	SLC39A11	Y	Y		yes
E01	chr7	133,039,961	+	EXOC4	chr7	133,040,558	+	EXOC4	N	Y	597	
E01	chr7	151,552,174	+	PRKAG2	chr7	151,552,718	+	PRKAG2	N	Y	544	
E01	chr9	131,556,898	+	TBC1D13	chr9	131,557,883	+	TBC1D13	N	Y	985	
E01	chr10	123,827,180	+	TACC2	chr10	123,831,512	+	TACC2	N	Y	4,332	
E01	chr12	18,222,218	+		chr12	18,234,138	-	RERGL	N	N	11,920	
E01	chr12	18,222,227	-		chr12	18,234,192	+	RERGL	N	N	11,965	
E01	chr12	51,376,811	+		chr12	51,380,334	+	SLC11A2	N	Y	3,523	
E01	chr12	53,595,988	+	ITGB7	chr12	53,596,590	+	ITGB7	N	Y	602	
E01	chr12	99,978,721	+	ANKS1B	chr12	99,982,702	+	ANKS1B	N	Y	3,981	
E01	chr16	4,067,093	+	ADCY9	chr16	4,067,573	+	ADCY9	N	Y	480	
E01	chr17	33,681,081	+	SLFN11	chr17	33,689,757	+	SLFN11	N	Y	8,676	
E01	chr17	33,687,392	+	SLFN11	chr17	33,689,759	+	SLFN11	N	Y	2,367	
E01	chr17	33,690,847	+	SLFN11	chr17	33,700,494	+	SLFN11	N	Y	9,647	
E01	chr17	71,562,137	-	SDK2	chr17	71,562,986	+	SDK2	N	N	849	
E01	chr18	24,134,023	+	KCTD1	chr18	24,134,444	+	KCTD1	N	Y	421	
E01	chrX	19,640,905	-	SH3KBP1	chrX	19,641,548	-	SH3KBP1	N	Y	643	
E01	chrX	32,931,076	+	DMD	chrX	32,931,604	+	DMD	N	Y	528	
F01	chr1	182,777,028	+	NPL	chr1	182,782,290	+	NPL	N	Y	5,262	
F01	chr2	28,720,813	+	PLB1	chr2	28,721,355	+	PLB1	N	Y	542	
F01	chr2	148,807,786	+	MBD5	chr2	148,813,762	+	MBD5	N	Y	5,976	
F01	chr3	61,827,238	+	PTPRG	chr3	61,837,175	+	PTPRG	N	Y	9,937	
F01	chr3	124,001,086	-	KALRN	chr18	75,652,754	-		Y	Y		yes
F01	chr3	173,240,733	+	NLGN1	chr3	173,241,712	+	NLGN1	N	Y	979	
F01	chr4	2,941,530	-	NOP14	chr12	16,970,231	-		Y	Y		yes
F01	chr4	2,941,851	+	NOP14	chr12	16,970,253	+		Y	Y		yes
F01	chr4	21,469,843	-	KCNIP4	chr4	110,248,224	+		N	N	88,778,381	yes

F01	chr4	169,165,016	-	DDX60	chr7	116,808,601	+	ST7	Y	N		yes
F01	chr4	189,013,485	+	TRIML2	chr4	189,015,126	+	TRIML2	N	Y	1,641	
F01	chr5	14,748,624	+	ANKH	chr5	14,750,271	+	ANKH	N	Y	1,647	
F01	chr5	14,749,156	-	ANKH	chr5	14,753,376	+	ANKH	N	N	4,220	
F01	chr5	14,749,156	-	ANKH	chr5	14,753,376	+	ANKH	N	N	4,220	
F01	chr5	14,749,513	+	ANKH	chr5	18,803,738	+		N	Y	4,054,225	yes
F01	chr5	14,749,546	-	ANKH	chr5	14,753,361	-	ANKH	N	Y	3,815	
F01	chr5	14,750,143	+	ANKH	chr13	106,513,997	-		Y	N		yes
F01	chr5	14,751,570	-	ANKH	chr5	14,751,898	+	ANKH	N	N	328	
F01	chr5	14,753,888	+	ANKH	chr5	14,753,923	-	ANKH	N	N	35	
F01	chr5	18,065,378	-		chr5	41,921,426	-	C5orf51	N	Y	23,856,048	yes
F01	chr5	18,861,724	-		chr5	41,792,602	-	OXCT1	N	Y	22,930,878	yes
F01	chr5	18,888,418	+		chr5	41,071,123	+	HEATR7B2	N	Y	22,182,705	yes
F01	chr5	28,352,392	-		chr5	41,343,636	+	PLCXD3	N	N	12,991,244	yes
F01	chr5	28,364,401	+		chr5	41,831,861	+	OXCT1	N	Y	13,467,460	yes
F01	chr5	28,649,138	+		chr5	41,831,486	-	OXCT1	N	N	13,182,348	yes
F01	chr5	41,198,129	-	C6	chr5	41,873,572	+		N	N	675,443	yes
F01	chr5	41,334,175	+	PLCXD3	chr5	41,826,701	+	OXCT1	N	Y	492,526	
F01	chr5	41,805,101	+	OXCT1	chr5	41,862,874	-	OXCT1	N	N	57,773	
F01	chr6	4,928,004	+	CDYL	chr6	4,928,621	+	CDYL	N	Y	617	
F01	chr7	4,299,796	+	SDK1	chr7	4,300,538	+	SDK1	N	Y	742	
F01	chr7	103,266,122	+	RELN	chr7	103,268,124	+	RELN	N	Y	2,002	
F01	chr7	114,045,855	+	ZNF555	chr8	73,139,003	+		Y	Y		yes
F01	chr8	97,792,198	+	PGCP	chr8	97,792,593	+	PGCP	N	Y	395	
F01	chr9	80,003,450	+	VPS13A	chr9	80,007,848	-	VPS13A	N	N	4,398	
F01	chr9	80,003,452	-	VPS13A	chr9	80,007,849	+	VPS13A	N	N	4,397	
F01	chr9	113,667,456	-	LPAR1	chr9	113,669,157	-	LPAR1	N	Y	1,701	
F01	chr9	113,667,553	+	LPAR1	chr9	113,669,264	+	LPAR1	N	Y	1,711	
F01	chr9	138,960,180	+	NACC2	chr9	138,963,225	+	NACC2	N	Y	3,045	
F01	chr10	58,789,805	-		chr15	60,713,666	+	NARG2	Y	N		yes
F01	chr10		+		chr15		-	NARG2	Y	N		yes

		58,789,809				60,713,908						
F01	chr10	88,642,127	+	BMPR1A	chr10	88,642,670	+	BMPR1A	N	Y	543	
F01	chr11	108,026,918	+		chr11	108,122,545	+	ATM	N	Y	95,627	
F01	chr12	32,330,618	+	BICD1	chr12	32,335,819	+	BICD1	N	Y	5,201	
F01	chr13	44,958,987	+	SERP2	chr13	106,470,584	+		N	Y	61,511,597	yes
F01	chr13	52,719,211	-	NEK3	chr13	107,935,687	+	FAM155A	N	N	55,216,476	yes
F01	chr13	93,965,435	-	GPC6	chr13	112,524,234	-		N	Y	18,558,799	yes
F01	chr15	40,102,355	+	GPR176	chr15	40,104,131	+	GPR176	N	Y	1,776	
F01	chr17	5,270,843	+	RABEP1	chr17	5,271,336	+	RABEP1	N	Y	493	
F01	chr17	31,632,861	+	ACCN1	chr17	31,636,171	+	ACCN1	N	Y	3,310	
F01	chr17	44,887,353	+	WNT3	chr17	44,887,685	+	WNT3	N	Y	332	
F01	chr17	46,400,405	+	SKAP1	chr17	46,402,558	+	SKAP1	N	Y	2,153	
F01	chr18	9,284,974	+	ANKRD12	chr18	9,286,141	+		N	Y	1,167	
F01	chr19	4,291,820	+		chr19	4,292,423	+	TMIGD2	N	Y	603	
F01	chr19	53,477,404	+	ZNF702P	chr19	53,477,955	+	ZNF702P	N	Y	551	
F01	chrX	17,061,042	+	REPS2	chrX	17,063,314	+	REPS2	N	Y	2,272	
F01	chrX	19,860,650	+	SH3KBP1	chrX	19,861,144	-	SH3KBP1	N	N	494	
F01	chrX	19,860,751	-	SH3KBP1	chrX	19,861,300	+	SH3KBP1	N	N	549	

Large structural alterations involving one or more genes in each of the six SMZL index genomes are shown. Left genes are arbitrarily designated based on chromosomal position numerically less than right genes (e.g., chr1 < chr2). Distance between intrachromosomal lesions is given in base pairs between breakpoints. Also indicated are the structural alterations that are displayed in the corresponding genome diagrams in Figure 1.

Table S2. Novel gene alterations in SMZL identified by DNA sequencing

Chr	Start	Gene Symbol	Var	mRNA	Protein	Str	bp	Ref	Call	aa	ref	call	impact	Gen	All	No
chr1	1,391,191	ATAD3C	snp	NM_001039211.2	NP_001034300.2	+	1,454	C	T	153	R	C	MISSENSE	A01	2	2
chr1	1,392,531	ATAD3C	snp	NM_001039211.2	NP_001034300.2	+	1,707	C	T	237	A	V	MISSENSE	C01	2	2
chr1	16,386,440	FAM131C	snp	NM_182623.2	NP_872429.2	-	563	C	G	124	G	A	MISSENSE	A01	2	2
chr1	16,386,440	FAM131C	snp	NM_182623.2	NP_872429.2	-	563	C	G	124	G	A	MISSENSE	F01	2	2
chr1	24,434,518	MYOM3	snp	NM_152372.3	NP_689585.3	-	368	G	C	68	A	G	MISSENSE	F01	1	2
chr1	24,435,079	MYOM3	snp	NM_152372.3	NP_689585.3	-	209	G	A	15	P	L	MISSENSE	C01	1	2
chr1	25,880,438	LDLRAP1	snp	NM_015627.2	NP_056442.2	+	228	C	T	38	R	W	MISSENSE	D01	2	2
chr1	25,893,444	LDLRAP1	snp	NM_015627.2	NP_056442.2	+	1,002	A	G	296	S	G	MISSENSE	A01	2	2
chr1	27,684,749	MAP3K6	snp	NM_004672.3	NP_004663.3	-	3,085	G	A	945	P	L	MISSENSE	B01	1	2
chr1	27,687,677	MAP3K6	snp	NM_004672.3	NP_004663.3	-	2,050	A	G	600	L	P	MISSENSE	E01	2	2
chr1	32,201,438	BAI2	snp	NM_001703.2	NP_001694.2	-	3,609	C	T	1,085	V	I	MISSENSE	D01	2	2
chr1	32,201,438	BAI2	snp	NM_001703.2	NP_001694.2	-	3,609	C	T	1,085	V	I	MISSENSE	E01	2	2
chr1	32,673,513	IQCC	snp	NM_001160042.1	NP_001153514.1	+	1,518	G	C	490	R	T	MISSENSE	D01	1	2
chr1	32,673,513	IQCC	snp	NM_001160042.1	NP_001153514.1	+	1,518	G	C	490	R	T	MISSENSE	E01	1	2
chr1	39,798,518	MACF1	snp	NM_033044.2	NP_149033.2	+	1,709	G	C	526	E	Q	MISSENSE	D01	1	2
chr1	39,801,827	MACF1	snp	NM_033044.2	NP_149033.2	+	5,018	A	G	1,629	T	A	MISSENSE	A01	2	2
chr1	39,853,686	MACF1	snp	NM_012090.3	NP_036222.3	+	9,191	T	A	2,995	V	E	MISSENSE	D01	1	2
chr1	52,704,152	ZFYVE9	snp	NM_007323.1	NP_015562.1	+	1,235	G	A	354	C	Y	MISSENSE	A01	1	2
chr1	52,732,445	ZFYVE9	snp	NM_004799.2	NP_004790.2	+	2,569	T	G	799	S	A	MISSENSE	A01	1	2

chr1	52,800,416	ZFYVE9	snp	NM_004799.2	NP_004790.2	+	3,818	T	G	1,215	L	R	MISSENSE	C01	1	2
chr1	78,276,648	LOC653631	snp	XM_003118959.1	XP_003119007.1	-	817	A	G	272	I	T	MISSENSE	F01	2	2
chr1	78,277,380	LOC653631	del	XM_003118959.1	XP_003119007.1	-	85	A		28	V	G	FRAMESHIFT	A01	2	2
chr1	78,277,435	LOC653631	snp	XM_003118959.1	XP_003119007.1	-	30	G	A	10	R	C	MISSENSE	A01	1	2
chr1	85,537,630	WDR63	snp	NM_145172.3	NP_660155.2	+	197	C	G	2	P	A	MISSENSE	B01	2	2
chr1	85,537,630	WDR63	snp	NM_145172.3	NP_660155.2	+	197	C	G	2	P	A	MISSENSE	F01	2	2
chr1	86,171,944	ZNHIT6	snp	NM_001170670.1	NP_001164141.1	-	847	C	G	232	M	I	MISSENSE	D01	2	2
chr1	86,173,773	ZNHIT6	snp	NM_001170670.1	NP_001164141.1	-	342	C	A	64	G	V	MISSENSE	F01	1	2
chr1	103,354,134	COL11A1	snp	NM_001854.3	NP_001845.3	-	4,923	G	C	1,535	P	A	MISSENSE	F01	2	2
chr1	103,354,471	COL11A1	snp	NM_001854.3	NP_001845.3	-	4,785	T	C	1,489	I	V	MISSENSE	A01	1	2
chr1	120,458,146	NOTCH2	snp	NM_024408.2	NP_077719.2	-	7,453	G	A	2,399	R	*	NONSENSE	B01	1	3
chr1	120,458,146	NOTCH2	snp	NM_024408.2	NP_077719.2	-	7,453	G	A	2,399	R	*	NONSENSE	C01	1	3
chr1	120,458,441	NOTCH2	ins	NM_024408.2	NP_077719.2	-	7,159		G	2,303	I	H	FRAMESHIFT	A01	2	3
chr1	152,129,252	RPTN	snp	NM_001122965.1	NP_001116437.1	-	386	C	T	107	A	T	MISSENSE	C01	2	2
chr1	152,129,252	RPTN	snp	NM_001122965.1	NP_001116437.1	-	386	C	T	107	A	T	MISSENSE	F01	2	2
chr1	152,276,796	FLG	sub	NM_002016.1	NP_002007.1	-	10,596	CCGGA	ACTGC	3,520	SG	AV	MISSENSE	D01	1	2
chr1	152,277,127	FLG	snp	NM_002016.1	NP_002007.1	-	10,269	C	T	3,411	G	R	MISSENSE	D01	2	2
chr1	152,277,136	FLG	snp	NM_002016.1	NP_002007.1	-	10,260	G	C	3,408	R	G	MISSENSE	D01	2	2
chr1	152,277,148	FLG	sub	NM_002016.1	NP_002007.1	-	10,248	T	GC	3,404	T	A	FRAMESHIFT	D01	2	2
chr1	152,283,645	FLG	snp	NM_002016.1	NP_002007.1	-	3,751	T	G	1,238	E	A	MISSENSE	A01	1	2
chr1	158,607,863	SPTA1	snp	NM_003126.2	NP_003117.2	-	5,346	G	T	1,715	F	L	MISSENSE	C01	2	2
chr1	158,614,183	SPTA1	snp	NM_003126.2	NP_003117.2	-	4,395	C	T	1,398	M	I	MISSENSE	B01	2	2

chr1	183,192,389	LAMC2	snp	NM_018891.2	NP_061486.2	+	1,197	G	A	294	R	Q	MISSENSE	C01	1	2
chr1	183,205,707	LAMC2	snp	NM_018891.2	NP_061486.2	+	2,883	G	C	856	R	P	MISSENSE	F01	1	2
chr1	190,067,734	FAM5C	snp	NM_199051.1	NP_950252.1	-	1,945	A	C	571	Y	D	MISSENSE	D01	2	2
chr1	190,423,815	FAM5C	snp	NM_199051.1	NP_950252.1	-	436	G	A	68	Q	*	NONSENSE	F01	1	2
chr1	207,133,092	FCAMR	snp	NM_001170631.1	NP_001164102.1	-	2,003	T	C	501	T	A	MISSENSE	F01	1	2
chr1	207,140,446	FCAMR	snp	NM_001170631.1	NP_001164102.1	-	618	C	T	39	R	K	MISSENSE	E01	2	2
chr1	211,526,768	TRAF5	snp	NM_004619.3	NP_004610.1	+	247	G	A	62	R	H	MISSENSE	A01	1	2
chr1	211,538,724	TRAF5	snp	NM_004619.3	NP_004610.1	+	763	G	A	234	R	Q	MISSENSE	D01	1	2
chr1	214,556,835	PTPN14	ins	NM_005401.4	NP_005392.2	-	3,015		G	788	I	H	FRAMESHIFT	A01	2	2
chr1	214,557,944	PTPN14	snp	NM_005401.4	NP_005392.2	-	1,905	C	T	417	S	N	MISSENSE	F01	2	2
chr1	215,901,703	USH2A	snp	NM_206933.2	NP_996816.2	-	12,120	C	T	3,911	E	K	MISSENSE	B01	2	2
chr1	215,916,662	USH2A	snp	NM_206933.2	NP_996816.2	-	11,790	C	T	3,801	E	K	MISSENSE	B01	2	2
chr1	216,172,287	USH2A	snp	NM_206933.2	NP_996816.2	-	6,984	A	G	2,199	F	L	MISSENSE	F01	2	2
chr1	225,380,397	DNAH14	snp	NM_001373.1	NP_001364.1	+	5,819	A	G	1,868	I	V	MISSENSE	A01	2	2
chr1	225,391,953	DNAH14	del	NM_001373.1	NP_001364.1	+	6,104	GTT		1,963	V		DELETE	A01	1	2
chr1	225,428,344	DNAH14	ins	NM_001373.1	NP_001364.1	+	6,717		A	2,167	I	N	FRAMESHIFT	A01	1	2
chr1	225,569,093	DNAH14	snp	NM_001373.1	NP_001364.1	+	12,798	C	T	4,194	P	L	MISSENSE	A01	2	2
chr1	225,569,141	DNAH14	snp	NM_001373.1	NP_001364.1	+	12,846	G	T	4,210	R	L	MISSENSE	F01	2	2
chr1	228,400,210	OBSCN	snp	NM_052843.2	NP_443075.2	+	770	G	C	242	G	R	MISSENSE	E01	1	3
chr1	228,456,381	OBSCN	snp	NM_052843.2	NP_443075.2	+	5,056	A	C	1,670	K	N	MISSENSE	D01	2	3
chr1	228,471,311	OBSCN	snp	NM_052843.2	NP_443075.2	+	8,889	G	A	2,948	R	H	MISSENSE	A01	1	3
chr1	228,559,449	OBSCN	snp	NM_001098623.1	NP_001092093.1	+	21,014	G	A	6,990	G	R	MISSENSE	E01	1	3

chr1	234,556,461	TARBP1	snp	NM_005646.3	NP_005637.3	-	3,540	G	C	1,180	P	A	MISSENSE	B01	1	2
chr1	234,565,853	TARBP1	snp	NM_005646.3	NP_005637.3	-	2,587	G	A	862	A	V	MISSENSE	C01	1	2
chr1	237,054,448	MTR	snp	NM_000254.2	NP_000245.2	+	3,446	G	C	1,007	K	N	MISSENSE	C01	1	2
chr1	237,058,681	MTR	snp	NM_000254.2	NP_000245.2	+	3,852	A	G	1,143	R	G	MISSENSE	A01	2	2
chr1	237,754,278	RYR2	snp	NM_001035.2	NP_001026.2	+	4,266	C	T	1,382	R	C	MISSENSE	C01	2	3
chr1	237,870,358	RYR2	snp	NM_001035.2	NP_001026.2	+	9,810	A	G	3,230	M	V	MISSENSE	F01	2	3
chr1	237,880,498	RYR2	snp	NM_001035.2	NP_001026.2	+	10,444	C	T	3,441	A	V	MISSENSE	D01	2	3
chr1	248,004,388	OR11L1	snp	NM_001001959.1	NP_001001959.1	-	809	G	C	269	I	M	MISSENSE	A01	1	2
chr1	248,004,875	OR11L1	sub	NM_001001959.1	NP_001001959.1	-	321	CC	TT	107	G	N	MISSENSE	B01	2	2
chr2	28,748,164	PLB1	snp	NM_153021.4	NP_694566.4	+	318	A	G	91	K	R	MISSENSE	B01	2	2
chr2	28,805,358	PLB1	snp	NM_153021.4	NP_694566.4	+	1,763	A	G	573	R	G	MISSENSE	E01	2	2
chr2	31,595,153	XDH	snp	NM_000379.3	NP_000370.2	-	1,874	C	T	598	R	H	MISSENSE	A01	2	2
chr2	31,621,522	XDH	snp	NM_000379.3	NP_000370.2	-	427	T	A	116	T	S	MISSENSE	E01	1	2
chr2	84,869,071	DNAH6	snp	NM_001370.1	NP_001361.1	+	5,108	T	A	1,657	S	T	MISSENSE	A01	1	2
chr2	85,012,832	DNAH6	ins	NM_001370.1	NP_001361.1	+	11,189		A	3,684	W	M	FRAMESHIFT	E01	1	2
chr2	86,292,463	POLR1A	snp	NM_015425.3	NP_056240.2	-	2,369	T	C	663	K	R	MISSENSE	F01	1	2
chr2	86,310,210	POLR1A	snp	NM_015425.3	NP_056240.2	-	1,189	T	C	270	N	D	MISSENSE	C01	1	2
chr2	111,806,831	ACOXL	snp	NM_001142807.1	NP_001136279.1	+	1,630	T	A	468	C	*	NONSENSE	A01	1	2
chr2	111,875,378	ACOXL	snp	NM_001142807.1	NP_001136279.1	+	1,952	G	T	576	G	*	NONSENSE	C01	2	2
chr2	127,646,858	LOC339760	del	XM_295058.6	XP_295058.6	+	86	C		29	P	Q	FRAMESHIFT	E01	1	2
chr2	127,656,552	LOC339760	snp	XM_295058.6	XP_295058.6	+	907	G	A	302	R	H	MISSENSE	D01	1	2
chr2	170,009,389	LRP2	snp	NM_004525.2	NP_004516.2	-	12,592	C	T	4,126	R	H	MISSENSE	A01	2	2

chr2	170,009,390	LRP2	snp	NM_004525.2	NP_004516.2	-	12,591	G	T	4,126	R	S	MISSENSE	C01	2	2
chr2	170,059,342	LRP2	snp	NM_004525.2	NP_004516.2	-	8,344	C	T	2,710	R	H	MISSENSE	A01	2	2
chr2	170,062,076	LRP2	snp	NM_004525.2	NP_004516.2	-	7,839	C	T	2,542	V	I	MISSENSE	A01	2	2
chr2	170,100,010	LRP2	snp	NM_004525.2	NP_004516.2	-	3,664	G	A	1,150	P	L	MISSENSE	A01	1	2
chr2	179,415,951	TTN	snp	NM_133432.3	NP_597676.3	-	64,710	G	A	21,495	R	W	MISSENSE	A01	1	4
chr2	179,437,626	TTN	snp	NM_133432.3	NP_597676.3	-	46,636	G	C	15,470	A	G	MISSENSE	D01	1	4
chr2	179,468,994	TTN	snp	NM_133432.3	NP_597676.3	-	27,823	C	T	9,199	R	Q	MISSENSE	A01	2	4
chr2	179,484,789	TTN	snp	NM_133432.3	NP_597676.3	-	19,758	A	C	6,511	C	G	MISSENSE	A01	2	4
chr2	179,586,813	TTN	snp	NM_133378.4	NP_596869.4	-	19,068	C	A	6,281	V	F	MISSENSE	B01	1	4
chr2	179,588,278	TTN	snp	NM_133378.4	NP_596869.4	-	18,040	C	T	5,938	C	Y	MISSENSE	F01	2	4
chr2	192,700,734	SDPR	snp	NM_004657.5	NP_004648.1	-	1,546	T	C	397	S	G	MISSENSE	C01	1	2
chr2	192,701,300	SDPR	snp	NM_004657.5	NP_004648.1	-	980	G	T	208	P	H	MISSENSE	D01	2	2
chr2	196,602,849	DNAH7	snp	NM_018897.2	NP_061720.2	-	11,970	A	C	3,956	M	R	MISSENSE	B01	2	2
chr2	196,865,506	DNAH7	snp	NM_018897.2	NP_061720.2	-	1,374	A	G	424	I	T	MISSENSE	C01	2	2
chr2	198,436,770	RFTN2	snp	NM_144629.2	NP_653230.2	-	1,868	A	C	488	F	L	MISSENSE	B01	2	2
chr2	198,508,888	RFTN2	snp	NM_144629.2	NP_653230.2	-	832	A	G	143	I	T	MISSENSE	C01	2	2
chr2	219,509,253	ZNF142	snp	NM_001105537.1	NP_001099007.1	-	2,420	T	A	661	E	V	MISSENSE	A01	1	2
chr2	219,509,284	ZNF142	snp	NM_001105537.1	NP_001099007.1	-	2,389	A	C	651	L	V	MISSENSE	D01	2	2
chr2	233,407,739	CHRNG	del	NM_005199.4	NP_005190.4	+	773	CT		250	PC	PC	FRAMESHIFT	B01	2	2
chr2	233,409,187	CHRNG	snp	NM_005199.4	NP_005190.4	+	1,167	A	G	382	I	V	MISSENSE	C01	2	2
chr2	238,242,175	COL6A3	snp	NM_057166.4	NP_476507.3	-	7,708	G	C	2,474	P	R	MISSENSE	A01	1	2
chr2	238,244,883	COL6A3	ins	NM_057166.4	NP_476507.3	-	7,323		G	2,348	A	R	FRAMESHIFT	E01	1	2

chr3	14,552,971	GRIP2	snp	NM_001080423.2	NP_001073892.2	-	2,026	C	T	675	R	Q	MISSENSE	D01	2	3
chr3	14,558,606	GRIP2	snp	NM_001080423.2	NP_001073892.2	-	1,561	C	T	520	R	Q	MISSENSE	F01	2	3
chr3	14,581,789	GRIP2	snp	NM_001080423.2	NP_001073892.2	-	316	G	A	105	T	I	MISSENSE	C01	1	3
chr3	28,476,603	ZCWPW2	snp	NM_001040432.1	NP_001035522.1	+	477	G	C	111	W	C	MISSENSE	F01	1	2
chr3	28,566,125	ZCWPW2	snp	NM_001040432.1	NP_001035522.1	+	1,159	G	A	339	E	K	MISSENSE	D01	1	2
chr3	41,831,232	ULK4	sub	NM_017886.2	NP_060356.2	-	2,312	TG	AA	703	CK	C*	NONSENSE	D01	1	2
chr3	41,949,347	ULK4	snp	NM_017886.2	NP_060356.2	-	1,371	G	A	390	P	S	MISSENSE	B01	1	2
chr3	45,972,662	FYCO1	snp	NM_024513.2	NP_078789.2	-	4,356	G	C	1,383	A	G	MISSENSE	B01	1	2
chr3	46,008,479	FYCO1	snp	NM_024513.2	NP_078789.2	-	2,551	C	G	781	Q	H	MISSENSE	D01	2	2
chr3	47,038,811	NBEAL2	snp	NM_015175.1	NP_055990.1	+	2,891	C	T	904	Q	*	NONSENSE	E01	2	2
chr3	47,049,614	NBEAL2	snp	NM_015175.1	NP_055990.1	+	7,836	G	A	2,552	G	E	MISSENSE	D01	1	2
chr3	47,447,413	PTPN23	snp	NM_015466.2	NP_056281.1	+	519	G	T	141	V	F	MISSENSE	C01	2	2
chr3	47,453,371	PTPN23	snp	NM_015466.2	NP_056281.1	+	4,083	G	A	1,329	V	M	MISSENSE	C01	1	2
chr3	47,454,418	PTPN23	snp	NM_015466.2	NP_056281.1	+	4,750	T	C	1,551	L	P	MISSENSE	E01	1	2
chr3	49,200,658	CCDC71	snp	NM_022903.3	NP_075054.3	-	1,120	T	A	327	Q	L	MISSENSE	D01	2	2
chr3	49,201,322	CCDC71	snp	NM_022903.3	NP_075054.3	-	456	T	C	106	M	V	MISSENSE	A01	1	2
chr3	97,887,849	OR5H15	del	NM_001005515.1	NP_001005515.1	+	306	T		102	S	P	FRAMESHIFT	C01	1	2
chr3	97,887,969	OR5H15	snp	NM_001005515.1	NP_001005515.1	+	426	C	T	142	R	W	MISSENSE	D01	2	2
chr3	121,415,713	GOLGB1	snp	NM_004487.3	NP_004478.3	-	3,766	C	G	1,213	R	P	MISSENSE	B01	2	2
chr3	121,417,232	GOLGB1	sub	NM_004487.3	NP_004478.3	-	2,245	CAT	TC	706	YE	*	NONSENSE	D01	2	2
chr3	142,083,977	XRN1	snp	NM_019001.3	NP_061874.3	-	3,371	T	C	1,101	E	G	MISSENSE	A01	1	2
chr3	142,095,387	XRN1	snp	NM_019001.3	NP_061874.3	-	2,830	C	T	921	G	R	MISSENSE	B01	2	2

chr3	150,282,123	EIF2A	snp	NM_032025.3	NP_114414.2	+	446	G	A	143	R	H	MISSENSE	A01	1	3
chr3	150,289,789	EIF2A	snp	NM_032025.3	NP_114414.2	+	872	C	G	285	T	S	MISSENSE	F01	2	3
chr3	150,301,670	EIF2A	del	NM_032025.3	NP_114414.2	+	1,746	GCTGGA		576	EL		DELETE	E01	1	3
chr3	156,396,122	TIPARP	snp	NM_015508.4	NP_056323.2	+	1,058	G	A	212	D	N	MISSENSE	B01	1	2
chr3	156,422,509	TIPARP	snp	NM_015508.4	NP_056323.2	+	1,985	G	A	521	D	N	MISSENSE	A01	1	2
chr3	193,185,127	ATP13A4	snp	NM_032279.2	NP_115655.2	-	1,198	A	G	363	V	A	MISSENSE	F01	2	2
chr3	193,210,748	ATP13A4	snp	NM_032279.2	NP_115655.2	-	597	G	T	163	Q	K	MISSENSE	C01	2	2
chr4	37,446,526	KIAA1239	snp	NM_001144990.1	NP_001138462.1	+	2,916	A	T	972	I	F	MISSENSE	D01	2	2
chr4	37,448,155	KIAA1239	snp	NM_001144990.1	NP_001138462.1	+	4,545	C	T	1,515	R	C	MISSENSE	C01	2	2
chr4	77,661,568	SHROOM3	snp	NM_020859.3	NP_065910.3	+	3,195	C	T	747	P	L	MISSENSE	E01	2	2
chr4	77,662,360	SHROOM3	snp	NM_020859.3	NP_065910.3	+	3,987	C	A	1,011	T	N	MISSENSE	B01	1	2
chr4	90,171,112	GPRIN3	snp	NM_198281.2	NP_938022.2	-	667	G	A	49	A	V	MISSENSE	E01	1	2
chr4	90,171,112	GPRIN3	snp	NM_198281.2	NP_938022.2	-	667	G	A	49	A	V	MISSENSE	F01	1	2
chr4	93,225,867	GRID2	snp	NM_001510.2	NP_001501.2	+	318	G	A	20	A	T	MISSENSE	B01	1	2
chr4	94,138,009	GRID2	snp	NM_001510.2	NP_001501.2	+	1,168	G	A	303	R	Q	MISSENSE	D01	1	2
chr4	94,411,796	GRID2	snp	NM_001510.2	NP_001501.2	+	2,123	A	T	621	E	D	MISSENSE	D01	2	2
chr4	120,327,472	LOC100128460	snp	XM_003118836.1	XP_003118884.1	-	1,495	G	T	498	A	D	MISSENSE	B01	2	2
chr4	120,327,752	LOC100128460	snp	XM_003118836.1	XP_003118884.1	-	1,215	C	T	405	G	S	MISSENSE	B01	2	2
chr4	120,327,982	LOC100128460	snp	XM_003118836.1	XP_003118884.1	-	985	G	A	328	P	L	MISSENSE	A01	1	2
chr5	13,900,378	DNAH5	snp	NM_001369.2	NP_001360.1	-	2,236	G	A	731	S	F	MISSENSE	F01	1	3
chr5	13,902,244	DNAH5	snp	NM_001369.2	NP_001360.1	-	1,688	G	C	548	N	K	MISSENSE	A01	2	3
chr5	13,922,301	DNAH5	snp	NM_001369.2	NP_001360.1	-	615	C	T	191	A	T	MISSENSE	B01	2	3

chr5	23,524,515	PRDM9	snp	NM_020227.2	NP_064612.2	+	1,205	A	G	341	R	G	MISSENSE	B01	2	2
chr5	23,526,914	PRDM9	snp	NM_020227.2	NP_064612.2	+	1,899	T	G	572	I	R	MISSENSE	E01	1	2
chr5	64,976,538	SGTB	snp	NM_019072.2	NP_061945.1	-	781	A	T	187	S	T	MISSENSE	B01	2	2
chr5	64,976,538	SGTB	snp	NM_019072.2	NP_061945.1	-	781	A	T	187	S	T	MISSENSE	F01	2	2
chr5	79,028,011	CMYA5	ins	NM_153610.3	NP_705838.3	+	3,495		AAAGGGAG	1,141	ASS	KGR	FRAMESHIFT	C01	1	2
chr5	79,033,120	CMYA5	snp	NM_153610.3	NP_705838.3	+	8,604	A	G	2,844	R	G	MISSENSE	A01	2	2
chr5	90,025,479	GPR98	snp	NM_032119.3	NP_115495.3	+	10,543	T	G	3,482	I	S	MISSENSE	B01	1	3
chr5	90,074,707	GPR98	snp	NM_032119.3	NP_115495.3	+	12,971	T	G	4,291	S	R	MISSENSE	E01	2	3
chr5	90,151,697	GPR98	snp	NM_032119.3	NP_115495.3	+	17,830	C	G	5,911	S	C	MISSENSE	A01	2	3
chr5	135,286,953	LECT2	snp	NM_002302.2	NP_002293.2	-	447	T	C	82	N	D	MISSENSE	C01	1	2
chr5	135,290,476	LECT2	snp	NM_002302.2	NP_002293.2	-	246	C	T	15	A	T	MISSENSE	E01	2	2
chr5	140,579,737	PCDHB11	ins	NM_018931.2	NP_061754.1	+	390		T	130	P	S	FRAMESHIFT	C01	2	2
chr5	140,581,711	PCDHB11	snp	NM_018931.2	NP_061754.1	+	2,364	T	C	788	F	L	MISSENSE	F01	1	2
chr5	140,769,005	PCDHGB4	snp	NM_032098.1	NP_115269.1	+	1,554	G	T	518	D	Y	MISSENSE	C01	2	2
chr5	140,769,007	PCDHGB4	del	NM_032098.1	NP_115269.1	+	1,556	CCACGAGCAGCTGCGCGCCTTCGA		518	DHEQLRAF		DELETE	A01	1	2
chr5	140,778,759	PCDHGB5	snp	NM_032099.1	NP_115270.1	+	1,065	G	A	355	A	T	MISSENSE	F01	1	2
chr5	140,779,717	PCDHGB5	snp	NM_032099.1	NP_115270.1	+	2,023	C	G	674	P	R	MISSENSE	A01	2	2
chr5	140,864,783	PCDHGC4	snp	NM_032406.1	NP_115782.1	+	43	C	T	14	A	V	MISSENSE	F01	2	2
chr5	140,866,831	PCDHGC4	snp	NM_032406.1	NP_115782.1	+	2,091	G	C	697	V	L	MISSENSE	B01	1	2
chr5	147,494,000	SPINK5	snp	NM_001127699.1	NP_001121171.1	+	2,036	G	A	654	G	D	MISSENSE	D01	1	2
chr5	147,513,363	SPINK5	snp	NM_006846.3	NP_006837.2	+	3,172	C	T	1,033	R	C	MISSENSE	B01	2	2
ch	149,447,8	CSF1R	sub	NM_005211.3	NP_005202.2	-	1,88	CAGGA	AGCAG	532	LL	PA	MISSENSE	D0	1	3

r5	01						9							1		
chr5	149,460,337	CSF1R	snp	NM_005211.3	NP_005202.2	-	590	T	C	99	Y	C	MISSENSE	B01	1	3
chr5	149,460,541	CSF1R	snp	NM_005211.3	NP_005202.2	-	386	A	C	31	V	G	MISSENSE	C01	2	3
chr5	149,754,225	TCOF1	snp	NM_001008657.2	NP_001008657.1	+	1,237	C	T	376	P	L	MISSENSE	A01	2	2
chr5	149,754,711	TCOF1	snp	NM_001008657.2	NP_001008657.1	+	1,581	A	G	491	M	V	MISSENSE	B01	2	2
chr5	150,422,227	TNIP1	snp	NM_006058.3	NP_006049.3	-	1,595	G	A	335	T	I	MISSENSE	D01	1	2
chr5	150,436,331	TNIP1	snp	NM_006058.3	NP_006049.3	-	1,210	G	C	207	L	V	MISSENSE	A01	1	2
chr5	176,815,129	SLC34A1	sub	NM_001167579.1	NP_001161051.1	+	883	CC	GG	259	GR	GG	MISSENSE	E01	1	2
chr5	176,816,712	SLC34A1	del	NM_001167579.1	NP_001161051.1	+	1,113	C		336	P	L	FRAMESHIFT	A01	2	2
chr6	4,068,949	C6orf146	snp	NM_173563.1	NP_775834.1	-	1,846	A	G	502	S	P	MISSENSE	E01	2	2
chr6	4,077,618	C6orf146	snp	NM_173563.1	NP_775834.1	-	369	G	C	9	N	K	MISSENSE	A01	1	2
chr6	7,232,011	RREB1	snp	NM_001168344.1	NP_001161816.1	+	4,135	C	T	1,226	P	L	MISSENSE	A01	2	2
chr6	7,247,037	RREB1	snp	NM_001168344.1	NP_001161816.1	+	4,645	C	T	1,396	T	I	MISSENSE	D01	2	2
chr6	7,576,579	DSP	snp	NM_001008844.1	NP_001008844.1	+	2,962	A	G	894	Y	C	MISSENSE	A01	2	2
chr6	7,581,647	DSP	snp	NM_004415.2	NP_004406.2	+	5,503	A	C	1,741	D	A	MISSENSE	E01	2	2
chr6	38,840,914	DNAH8	snp	NM_001371.2	NP_001362.2	+	7,419	A	G	2,273	I	V	MISSENSE	A01	2	2
chr6	38,917,230	DNAH8	snp	NM_001371.2	NP_001362.2	+	12,081	T	A	3,827	S	T	MISSENSE	A01	1	2
chr6	38,998,102	DNAH8	snp	NM_001371.2	NP_001362.2	+	14,007	C	T	4,469	R	*	NONSENSE	E01	2	2
chr6	44,442,032	LOC100128935	snp	XM_001725735.1	XP_001725787.1	+	244	T	G	81	V	G	MISSENSE	B01	1	2
chr6	44,442,040	LOC100128935	snp	XM_001725735.1	XP_001725787.1	+	252	A	G	84	R	G	MISSENSE	B01	1	2
chr6	44,442,044	LOC100128935	ins	XM_001725735.1	XP_001725787.1	+	256		TGGTGGCAGATCCTGGGGTCAGAGGGC	90		GVRGLVADP	INSERT	C01	2	2

chr6	56,480,499	DST	snp	NM_001723.5	NP_001714.1	-	7,872	T	C	2,588	I	V	MISSENSE	C01	1	3
chr6	56,480,499	DST	snp	NM_001723.5	NP_001714.1	-	7,872	T	C	2,588	I	V	MISSENSE	F01	1	3
chr6	56,482,922	DST	snp	NM_001723.5	NP_001714.1	-	6,016	A	G	1,969	F	S	MISSENSE	D01	2	3
chr6	64,694,369	EYS	snp	NM_001142800.1	NP_001136272.1	-	7,498	T	G	2,320	I	L	MISSENSE	A01	2	2
chr6	65,300,476	EYS	snp	NM_001142800.1	NP_001136272.1	-	5,820	A	T	1,760	Y	*	NONSENSE	E01	2	2
chr6	90,380,732	MDN1	snp	NM_014611.1	NP_055426.1	-	13,976	C	T	4,620	V	I	MISSENSE	F01	2	3
chr6	90,384,233	MDN1	ins	NM_014611.1	NP_055426.1	-	12,952		G	4,278	PQ	PS	FRAMESHIFT	A01	2	3
chr6	90,411,757	MDN1	snp	NM_014611.1	NP_055426.1	-	8,286	C	T	2,723	R	Q	MISSENSE	B01	2	3
chr6	99,848,777	SFRS18	snp	NM_032870.2	NP_116259.2	-	2,283	G	A	685	R	C	MISSENSE	B01	1	2
chr6	99,848,777	SFRS18	snp	NM_032870.2	NP_116259.2	-	2,283	G	A	685	R	C	MISSENSE	F01	1	2
chr6	112,450,155	LAMA4	snp	NM_001105206.1	NP_001098676.1	-	4,563	A	C	1,418	L	V	MISSENSE	C01	2	2
chr6	112,486,396	LAMA4	snp	NM_001105206.1	NP_001098676.1	-	1,941	G	A	544	R	C	MISSENSE	F01	1	2
chr6	152,501,415	SYNE1	snp	NM_015293.2	NP_056108.2	-	7,006	C	T	2,295	R	Q	MISSENSE	F01	2	2
chr6	152,806,103	SYNE1	del	NM_033071.2	NP_149062.1	-	1,508	A		357	FK	LS	FRAMESHIFT	C01	2	2
chr6	155,465,776	TIAM2	snp	NM_012454.3	NP_036586.2	+	1,875	T	A	555	N	K	MISSENSE	B01	1	2
chr6	155,577,713	TIAM2	sub	NM_012454.3	NP_036586.2	+	4,772	GCTT	CCCA	1,521	SL	TQ	MISSENSE	C01	1	2
chr6	170,169,737	C6orf70	snp	NM_018341.1	NP_060811.1	+	1,194	A	G	387	N	D	MISSENSE	D01	2	2
chr6	170,176,120	C6orf70	snp	NM_018341.1	NP_060811.1	+	1,512	C	G	493	R	G	MISSENSE	E01	2	2
chr7	30,818,141	FAM188B	snp	NM_032222.2	NP_115598.2	+	234	T	G	52	L	R	MISSENSE	D01	2	2
chr7	30,868,298	FAM188B	snp	NM_032222.2	NP_115598.2	+	1,154	A	G	359	N	D	MISSENSE	B01	2	2
chr7	47,840,309	PKD1L1	snp	NM_138295.2	NP_612152.1	-	8,129	C	G	2,709	Q	H	MISSENSE	D01	2	3
chr7	47,955,055	PKD1L1	snp	NM_138295.2	NP_612152.1	-	1,200	A	T	400	Y	N	MISSENSE	E01	2	3

chr7	47,970,806	PKD1L1	snp	NM_138295.2	NP_612152.1	-	630	C	T	210	G	R	MISSENSE	A01	2	3
chr7	95,814,275	SLC25A13	snp	NM_001160210.1	NP_001153682.1	-	1,174	C	A	327	E	D	MISSENSE	D01	1	2
chr7	95,906,623	SLC25A13	ins	NM_001160210.1	NP_001153682.1	-	287		A	33	FM	FH	FRAMESHIFT	C01	1	2
chr7	98,988,688	ARPC1B	snp	NM_005720.2	NP_005711.1	+	762	C	T	224	T	I	MISSENSE	A01	2	2
chr7	98,992,083	ARPC1B	snp	NM_005720.2	NP_005711.1	+	1,179	C	T	363	S	L	MISSENSE	B01	2	2
chr7	99,160,083	ZNF655	snp	NM_024061.3	NP_076966.1	+	708	A	G	105	M	V	MISSENSE	B01	2	2
chr7	99,160,083	ZNF655	snp	NM_024061.3	NP_076966.1	+	708	A	G	105	M	V	MISSENSE	C01	2	2
chr7	100,417,396	EPHB4	snp	NM_004444.4	NP_004435.3	-	1,569	C	T	359	R	H	MISSENSE	B01	2	2
chr7	100,421,431	EPHB4	snp	NM_004444.4	NP_004435.3	-	735	C	T	81	G	D	MISSENSE	C01	2	2
chr7	100,635,879	MUC12	snp	NM_001164462.1	NP_001157934.1	+	2,035	C	A	678	T	N	MISSENSE	D01	1	2
chr7	100,648,481	MUC12	snp	NM_001164462.1	NP_001157934.1	+	14,637	C	A	4,879	P	T	MISSENSE	F01	1	2
chr7	100,674,463	MUC17	snp	NM_001040105.1	NP_001035194.1	+	198	A	G	48	N	S	MISSENSE	A01	2	2
chr7	100,682,072	MUC17	snp	NM_001040105.1	NP_001035194.1	+	7,428	C	G	2,458	A	G	MISSENSE	E01	2	2
chr7	100,683,850	MUC17	snp	NM_001040105.1	NP_001035194.1	+	9,206	C	A	3,051	P	T	MISSENSE	E01	1	2
chr7	100,686,653	MUC17	ins	NM_001040105.1	NP_001035194.1	+	12,009		T	3,985	S	F	FRAMESHIFT	E01	2	2
chr7	107,674,717	LAMB4	snp	NM_007356.2	NP_031382.2	-	4,836	G	A	1,584	R	W	MISSENSE	A01	1	2
chr7	107,689,885	LAMB4	sub	NM_007356.2	NP_031382.2	-	4,088	GCA	CTT	1,334	SA	SR	MISSENSE	B01	1	2
chr7	111,379,197	DOCK4	snp	NM_014705.3	NP_055520.3	-	5,465	G	C	1,732	P	A	MISSENSE	D01	1	2
chr7	111,580,165	DOCK4	snp	NM_014705.3	NP_055520.3	-	1,244	T	C	325	M	V	MISSENSE	F01	2	2
chr7	121,944,238	FEZF1	snp	NM_001160264.1	NP_001153736.1	-	326	T	C	84	S	G	MISSENSE	B01	1	2
chr7	121,944,238	FEZF1	snp	NM_001160264.1	NP_001153736.1	-	326	T	C	84	S	G	MISSENSE	F01	1	2
chr7	150,174,649	GIMAP8	snp	NM_175571.2	NP_783161.1	+	2,109	T	C	593	C	R	MISSENSE	D01	1	2

chr7	150,174,812	GIMAP8	snp	NM_175571.2	NP_783161.1	+	2,272	A	C	647	Q	P	MISSENSE	A01	2	2
chr7	158,679,716	WDR60	snp	NM_018051.4	NP_060521.4	+	1,098	C	T	313	A	V	MISSENSE	D01	2	2
chr7	158,704,360	WDR60	ins	NM_018051.4	NP_060521.4	+	1,738		A	526	NT	KY	FRAMESHIFT	F01	1	2
chr8	11,400,820	BLK	snp	NM_001715.2	NP_001706.2	+	668	G	A	29	A	T	MISSENSE	A01	1	2
chr8	11,412,933	BLK	snp	NM_001715.2	NP_001706.2	+	1,293	G	A	237	R	Q	MISSENSE	D01	1	2
chr8	13,425,155	C8orf48	snp	NM_001007090.2	NP_001007091.1	+	804	A	C	218	K	T	MISSENSE	C01	2	2
chr8	13,425,155	C8orf48	snp	NM_001007090.2	NP_001007091.1	+	804	A	C	218	K	T	MISSENSE	E01	2	2
chr8	17,573,277	MTUS1	snp	NM_001001931.2	NP_001001931.1	-	642	T	C	107	K	R	MISSENSE	F01	1	2
chr8	17,611,797	MTUS1	snp	NM_001001924.2	NP_001001924.1	-	1,992	A	C	506	F	V	MISSENSE	C01	2	2
chr8	36,644,902	KCNU1	snp	NM_001031836.2	NP_001027006.2	+	361	A	G	91	E	G	MISSENSE	D01	2	2
chr8	36,780,142	KCNU1	snp	NM_001031836.2	NP_001027006.2	+	2,818	C	T	910	A	V	MISSENSE	C01	2	2
chr8	42,163,888	IKBKB	snp	NM_001190722.1	NP_001177651.1	+	691	A	C	168	Y	S	MISSENSE	F01	2	2
chr8	42,174,344	IKBKB	snp	NM_001190722.1	NP_001177651.1	+	1,233	G	T	349	D	Y	MISSENSE	B01	2	2
chr8	52,287,174	PXDNL	snp	NM_144651.4	NP_653252.3	-	3,774	C	T	1,224	R	Q	MISSENSE	D01	2	2
chr8	52,321,965	PXDNL	snp	NM_144651.4	NP_653252.3	-	2,318	G	A	739	P	S	MISSENSE	F01	1	2
chr8	110,393,639	PKHD1L1	snp	NM_177531.4	NP_803875.2	+	308	G	T	68	A	S	MISSENSE	C01	2	2
chr8	110,510,963	PKHD1L1	snp	NM_177531.4	NP_803875.2	+	10,895	A	C	3,597	S	R	MISSENSE	F01	2	2
chr8	144,942,299	EPPK1	snp	NM_031308.1	NP_112598.2	-	5,134	C	T	1,707	G	S	MISSENSE	B01	2	2
chr8	144,945,134	EPPK1	snp	NM_031308.1	NP_112598.2	-	2,299	C	A	762	D	Y	MISSENSE	E01	1	2
chr8	144,992,659	PLEC1	snp	NM_201384.1	NP_958786.1	-	11,457	C	T	3,776	E	K	MISSENSE	D01	2	2
chr8	145,004,372	PLEC1	snp	NM_201384.1	NP_958786.1	-	2,679	C	T	850	V	M	MISSENSE	A01	2	2
chr9	18,639,332	ADAMTSL1	snp	NM_052866.3	NP_443098.3	+	884	G	A	252	S	N	MISSENSE	D01	1	2

chr9	18,906,813	ADAMTSL1	snp	NM_001040272.4	NP_001035362.3	+	5,212	C	T	1,695	H	Y	MISSENSE	B01	2	2
chr9	35,906,543	HRCT1	snp	NM_001039792.1	NP_001034881.1	+	355	T	A	86	L	H	MISSENSE	D01	2	2
chr9	35,906,549	HRCT1	snp	NM_001039792.1	NP_001034881.1	+	361	A	C	88	H	P	MISSENSE	C01	2	2
chr9	86,554,607	C9orf64	snp	NM_032307.3	NP_115683.3	-	1,091	A	G	281	S	P	MISSENSE	C01	2	2
chr9	86,571,125	C9orf64	snp	NM_032307.3	NP_115683.3	-	537	C	T	96	S	N	MISSENSE	F01	2	2
chr9	113,163,240	SVEP1	snp	NM_153366.3	NP_699197.3	-	10,051	C	T	3,238	V	I	MISSENSE	C01	1	2
chr9	113,169,012	SVEP1	snp	NM_153366.3	NP_699197.3	-	9,203	G	A	2,955	A	V	MISSENSE	C01	1	2
chr9	113,205,962	SVEP1	snp	NM_153366.3	NP_699197.3	-	4,837	T	A	1,500	N	Y	MISSENSE	A01	1	2
chr9	116,191,186	C9orf43	snp	NM_152786.1	NP_689999.1	+	1,560	C	T	371	T	M	MISSENSE	B01	2	3
chr9	116,191,186	C9orf43	snp	NM_152786.1	NP_689999.1	+	1,560	C	T	371	T	M	MISSENSE	D01	2	3
chr9	116,191,186	C9orf43	snp	NM_152786.1	NP_689999.1	+	1,560	C	T	371	T	M	MISSENSE	E01	2	3
chr9	123,903,050	CEP110	snp	NM_007018.4	NP_008949.4	+	2,500	A	G	823	T	A	MISSENSE	E01	2	2
chr9	123,904,537	CEP110	snp	NM_007018.4	NP_008949.4	+	2,891	T	C	953	L	S	MISSENSE	C01	1	2
chr9	131,581,160	ENDOG	snp	NM_004435.2	NP_004426.2	+	382	G	C	65	K	N	MISSENSE	F01	1	2
chr9	131,584,883	ENDOG	del	NM_004435.2	NP_004426.2	+	1,073	AAGT		296	K*	?	NONSTOP	A01	2	2
chr9	133,799,637	FIBCD1	snp	NM_032843.4	NP_116232.3	-	942	C	T	232	R	Q	MISSENSE	B01	2	2
chr9	133,804,985	FIBCD1	snp	NM_032843.4	NP_116232.3	-	764	C	T	173	A	T	MISSENSE	A01	2	2
chr9	138,661,829	KCNT1	snp	NM_020822.2	NP_065873.2	+	1,621	G	A	515	M	I	MISSENSE	B01	1	2
chr9	138,678,153	KCNT1	snp	NM_020822.2	NP_065873.2	+	3,362	G	A	1,096	G	S	MISSENSE	D01	1	2
chr10	5,541,276	CALML5	snp	NM_017422.4	NP_059118.2	-	256	T	C	41	K	R	MISSENSE	B01	1	2
chr10	5,541,276	CALML5	snp	NM_017422.4	NP_059118.2	-	256	T	C	41	K	R	MISSENSE	D01	1	2

chr10	19,422,643	C10orf112	snp	XM_295865.8	XP_295865.8	+	705	A	C	235	T	P	MISSENSE	E01	2	2
chr10	19,678,529	C10orf112	snp	XM_295865.8	XP_295865.8	+	4,653	C	T	1,551	L	F	MISSENSE	B01	2	2
chr10	46,965,802	SYT15	snp	NM_181519.2	NP_852660.1	-	886	C	T	244	G	D	MISSENSE	B01	2	2
chr10	46,967,492	SYT15	snp	NM_181519.2	NP_852660.1	-	736	C	T	194	R	H	MISSENSE	D01	2	2
chr10	46,967,533	SYT15	snp	NM_181519.2	NP_852660.1	-	695	G	T	180	C	*	NONSENSE	B01	2	2
chr10	49,393,642	FRMPD2	snp	NM_001018071.3	NP_001018081.3	-	2,642	C	T	770	R	Q	MISSENSE	B01	2	2
chr10	49,430,387	FRMPD2	del	NM_001018071.3	NP_001018081.3	-	1,753	A		474	L	C	FRAMESHIFT	E01	2	2
chr10	49,431,224	FRMPD2	snp	NM_001018071.3	NP_001018081.3	-	1,587	C	A	418	M	I	MISSENSE	E01	1	2
chr10	49,457,149	FRMPD2	snp	NM_001018071.3	NP_001018081.3	-	553	G	C	74	R	G	MISSENSE	B01	1	2
chr10	76,789,773	MYST4	ins	NM_012330.2	NP_036462.2	+	5,684		GCTGCAGCAT	1,736	QTSIS	HAAAN	FRAMESHIFT	D01	2	2
chr10	76,789,778	MYST4	snp	NM_012330.2	NP_036462.2	+	5,689	A	T	1,732	S	C	MISSENSE	E01	1	2
chr10	115,374,673	NRAP	snp	NM_006175.3	NP_006166.3	-	3,168	C	T	1,001	R	Q	MISSENSE	E01	2	2
chr10	115,388,685	NRAP	snp	NM_006175.3	NP_006166.3	-	2,193	A	G	676	L	P	MISSENSE	A01	2	2
chr10	115,399,984	NRAP	snp	NM_006175.3	NP_006166.3	-	1,486	C	T	440		ATAAGT	DISRUPT	E01	2	2
chr1	127,677,219	FANK1	snp	NM_145235.3	NP_660278.3	+	395	C	T	97	P	S	MISSENSE	A01	2	2

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chr10	127,693,483	FANK1	del	NM_145235.3	NP_660278.3	+	674	GA		190	D	C	FRAMESHIFT	C01	2	2
chr11	1,078,564	MUC2	snp	NM_002457.2	NP_002448.2	+	799	C	T	257	P	L	MISSENSE	A01	2	2
chr11	1,096,513	MUC2	snp	NM_002457.2	NP_002448.2	+	13,651	G	A	4,541	R	H	MISSENSE	B01	1	2
chr11	1,266,598	MUC5B	snp	NM_002458.2	NP_002449.2	+	8,547	C	T	2,829	P	L	MISSENSE	F01	2	3
chr11	1,266,628	MUC5B	snp	NM_002458.2	NP_002449.2	+	8,577	C	T	2,839	T	M	MISSENSE	D01	2	3
chr11	1,270,317	MUC5B	ins	NM_002458.2	NP_002449.2	+	12,266		GTCCAGCCCT	4,069	HPSS	VQPS	FRAMESHIFT	F01	2	3
chr11	1,271,992	MUC5B	sub	NM_002458.2	NP_002449.2	+	13,941	CCACACCCACAACCAGT	GA	4,627	TTPTTS	R	DELETE+	A01	2	3
chr11	1,272,787	MUC5B	snp	NM_002458.2	NP_002449.2	+	14,736	C	T	4,892	T	M	MISSENSE	D01	2	3
chr11	6,555,243	DNHD1	snp	NM_144666.2	NP_653267.2	+	3,402	G	T	946	E	*	NONSENSE	C01	2	3
chr11	6,567,432	DNHD1	snp	NM_144666.2	NP_653267.2	+	5,827	G	A	1,754	R	H	MISSENSE	D01	1	3
chr11	6,568,232	DNHD1	snp	NM_144666.2	NP_653267.2	+	6,627	G	C	2,021	G	R	MISSENSE	A01	1	3
chr11	43,465,020	TTC17	snp	NM_018259.5	NP_060729.2	+	2,467	C	T	799	R	W	MISSENSE	C01	2	2
chr11	43,469,617	TTC17	snp	NM_018259.5	NP_060729.2	+	2,801	T	C	910	V	A	MISSENSE	D01	1	2
chr11	44,193,164	EXT2	snp	NM_207122.1	NP_997005.1	+	1,511	G	A	392	R	Q	MISSENSE	D01	1	2

chr11	44,254,029	EXT2	snp	NM_207122.1	NP_997005.1	+	2,123	C	T	596	A	V	MISSENSE	E01	2	2
chr11	57,070,275	TNKS1BP1	ins	NM_033396.2	NP_203754.2	-	4,505		CC	1,446	PP	PA	FRAMESHIFT	E01	1	2
chr11	57,080,676	TNKS1BP1	ins	NM_033396.2	NP_203754.2	-	1,650		GAGG	498	PI	LP	FRAMESHIFT	F01	2	2
chr11	57,088,127	TNKS1BP1	del	NM_033396.2	NP_203754.2	-	300	TGGCTTGGCAGGCAGGGC		49	KPALPA		DELETE	E01	1	2
chr11	62,289,320	AHNAK	snp	NM_001620.1	NP_001611.1	-	12,867	T	C	4,189	K	E	MISSENSE	B01	1	2
chr11	62,291,360	AHNAK	snp	NM_001620.1	NP_001611.1	-	10,827	T	C	3,509	M	V	MISSENSE	E01	1	2
chr11	89,075,266	NOX4	snp	NM_016931.3	NP_058627.1	-	1,550	A	C	437	F	V	MISSENSE	A01	2	3
chr11	89,075,266	NOX4	snp	NM_001143837.1	NP_001137309.1	-	1,775	A	C	413	F	V	MISSENSE	A01	2	3
chr11	89,182,665	NOX4	snp	NM_001143836.1	NP_001137308.1	-	529	C	A	96	L	F	MISSENSE	B01	1	3
chr11	89,223,714	NOX4	snp	NM_001143836.1	NP_001137308.1	-	302	A	T	21	W	R	MISSENSE	C01	2	3
chr11	92,533,557	FAT3	snp	NM_001008781.2	NP_001008781.2	+	7,395	G	A	2,459	R	Q	MISSENSE	B01	1	2
chr11	92,577,780	FAT3	snp	NM_001008781.2	NP_001008781.2	+	11,264	G	T	3,749	E	*	NONSENSE	C01	2	2
chr11	96,074,986	MAML2	snp	NM_032427.1	NP_115803.1	-	1,357	C	A	24	G	W	MISSENSE	D01	1	2
chr11	96,074,028	MAML2	snp	NM_032427.2	NP_115803.2	-	1,318	C	A	11	A	S	MISSENSE	N/A	1	2
chr1	108,122,589	ATM	snp	NM_000051.3	NP_000042.3	+	2,018	C	T	544	A	V	MISSENSE	E01	2	3

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chr11	108,122,603	ATM	snp	NM_000051.3	NP_000042.3	+	2,032	A	G	549	I	V	MISSENSE	A01	2	3
chr11	108,192,065	ATM	snp	NM_000051.3	NP_000042.3	+	6,875	A	G	2,163	E	G	MISSENSE	F01	2	3
chr11	117,308,648	DSCAML1	snp	NM_020693.2	NP_065744.2	-	4,575	C	T	1,524	R	H	MISSENSE	E01	2	2
chr11	117,395,611	DSCAML1	snp	NM_020693.2	NP_065744.2	-	1,026	C	T	341	S	N	MISSENSE	C01	2	2
chr12	10,862,594	CSDA	snp	NM_003651.4	NP_003642.3	-	934	T	G	230	Q	P	MISSENSE	E01	1	2
chr12	10,862,602	CSDA	del	NM_003651.4	NP_003642.3	-	915	ATACTGGGGGCG		225	PQYR		DELETE	C01	2	2
chr12	40,874,761	LOC100506072	snp	XM_003118944.1	XP_003118992.1	+	6,681	C	T	2,102	P	S	MISSENSE	B01	2	4
chr12	40,878,406	LOC100506072	snp	XM_003118944.1	XP_003118992.1	+	10,326	T	C	3,317	S	P	MISSENSE	B01	1	4
chr12	40,879,527	LOC100506072	ins	XM_003118944.1	XP_003118992.1	+	11,389		AAGTGACAGGGACAACTGGACCATTAGCTG	3,671		EVTGTTGPLA	INSERT	A01	1	4
chr12	40,883,583	LOC100506072	sub	XM_003118944.1	XP_003118992.1	+	15,445	AA	GG	5,023	E	G	MISSENSE	B01	1	4
chr12	40,883,643	LOC100506072	snp	XM_003118944.1	XP_003118992.1	+	15,505	A	G	5,043	E	G	MISSENSE	D01	2	4
chr12	40,883,643	LOC100506072	snp	XM_003118944.1	XP_003118992.1	+	15,505	A	G	5,043	E	G	MISSENSE	E01	2	4
chr12	40,883,646	LOC100506072	snp	XM_003118944.1	XP_003118992.1	+	15,508	T	C	5,044	I	T	MISSENSE	D01	2	4
chr1	40,884,664	LOC100506072	sub	XM_003118944.1	XP_003118992.1	+	16,526	AC	CAT	5,383	TP	TI	FRAMESHIFT	A01	2	4

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chr12	49,433,256	MLL2	del	NM_003482.3	NP_003473.3	-	8,171	CTCAAAGGCAGGGCTGCTG		2,725	SPAFEQL	*	NONSENSE	C01	2	3
chr12	49,434,698	MLL2	ins	NM_003482.3	NP_003473.3	-	6,854		T	2,285	E	R	FRAMESHIFT	A01	2	3
chr12	49,435,239	MLL2	snp	NM_003482.3	NP_003473.3	-	6,312	G	A	2,104	R	C	MISSENSE	D01	1	3
chr12	50,186,352	NCKAP5L	snp	NM_001037806.3	NP_001032895.2	-	3,869	C	A	1,222	G	V	MISSENSE	D01	1	2
chr12	50,190,188	NCKAP5L	snp	NM_001037806.3	NP_001032895.2	-	1,655	C	G	484	R	P	MISSENSE	E01	2	2
chr12	50,745,014	FAM186A	snp	NM_001145475.1	NP_001138947.1	-	5,737	G	A	1,866	S	F	MISSENSE	A01	1	2
chr12	50,747,312	FAM186A	snp	NM_001145475.1	NP_001138947.1	-	3,439	A	G	1,100	L	P	MISSENSE	E01	2	2
chr12	53,162,772	KRT76	del	NM_015848.4	NP_056932.2	-	1,692	ACT		546	S		DELETE	F01	1	2
chr12	53,170,959	KRT76	snp	NM_015848.4	NP_056932.2	-	169	G	C	38	A	G	MISSENSE	D01	1	2
chr12	111,893,920	ATXN2	ins	NM_002973.3	NP_002964.3	-	3,818		G	1,218	PA	PS	FRAMESHIFT	A01	2	2
chr12	111,902,513	ATXN2	snp	NM_002973.3	NP_002964.3	-	3,483	G	A	1,107	P	S	MISSENSE	D01	1	2
chr12	121,881,552	KDM2B	snp	NM_032590.4	NP_115979.3	-	2,598	G	A	831	S	L	MISSENSE	C01	1	2
chr12	122,018,760	KDM2B	snp	NM_032590.4	NP_115979.3	-	159	T	C	18	H	R	MISSENSE	B01	1	2
chr12	124,821,522	NCOR2	snp	NM_006312.4	NP_006303.3	-	6,241	G	C	1,973	S	C	MISSENSE	B01	1	3

chr12	124,856,829	NCOR2	snp	NM_006312.4	NP_006303.3	-	2,865	C	T	848	E	K	MISSENSE	B01	2	3
chr12	124,856,829	NCOR2	snp	NM_006312.4	NP_006303.3	-	2,865	C	T	848	E	K	MISSENSE	E01	2	3
chr12	124,885,169	NCOR2	snp	NM_006312.4	NP_006303.3	-	2,010	C	T	563	V	M	MISSENSE	D01	2	3
chr12	132,466,834	EP400	sub	NM_015409.3	NP_056224.2	+	1,846	CC	AT	579	P	M	MISSENSE	C01	1	2
chr12	132,466,834	EP400	sub	NM_015409.3	NP_056224.2	+	1,846	CC	AT	579	P	M	MISSENSE	D01	1	2
chr13	25,009,612	PARP4	snp	NM_006437.3	NP_006428.2	-	3,771	C	T	1,221		CTGTCTTCTGGGCAA	DISRUPT	F01	2	2
chr13	25,075,770	PARP4	snp	NM_006437.3	NP_006428.2	-	439	C	T	111	E	K	MISSENSE	A01	2	2
chr13	45,008,838	TSC22D1	snp	NM_183422.2	NP_904358.2	-	3,635	G	A	1,048	P	S	MISSENSE	D01	1	2
chr13	45,148,069	TSC22D1	snp	NM_183422.2	NP_904358.2	-	2,631	C	A	713	G	V	MISSENSE	D01	2	2
chr13	45,149,972	TSC22D1	snp	NM_183422.2	NP_904358.2	-	728	G	A	79	P	S	MISSENSE	C01	1	2
chr13	64,407,057	LOC100507505	del	XM_003118893.1	XP_003118941.1	-	45	ACCAGAGCCATAGCCACAGCCATA		21	SGYGCGYG		DELETE	C01	2	2
chr13	64,407,057	LOC100507505	del	XM_003118893.1	XP_003118941.1	-	45	ACCAGAGCCATAGCCACAGCCATA		21	SGYGCGYG		DELETE	F01	2	2
chr13	103,382,690	hCG_2011852	snp	NM_001146197.1	NP_001139669.1	-	20,495	A	G	6,785	C	R	MISSENSE	F01	2	3
chr13	103,383,357	hCG_2011852	snp	NM_001146197.1	NP_001139669.1	-	19,828	T	A	6,562	K	N	MISSENSE	A01	1	3
chr1	103,384,133	hCG_2011852	snp	NM_001146197.1	NP_001139669.1	-	19,052	G	A	6,304	Q	*	NONSENSE	E01	1	3

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chr13	103,393,784	hCG_2011852	snp	NM_001146197.1	NP_001139669.1	-	9,401	G	A	3,087	L	F	MISSENSE	A01	1	3
chr14	20,295,970	OR4N2	snp	NM_001004723.1	NP_001004723.1	+	363	C	T	121	R	C	MISSENSE	B01	2	2
chr14	20,295,970	OR4N2	snp	NM_001004723.1	NP_001004723.1	+	363	C	T	121	R	C	MISSENSE	F01	2	2
chr14	21,780,007	RPGRIP1	snp	NM_020366.3	NP_065099.3	+	955	A	G	318	H	R	MISSENSE	C01	2	2
chr14	21,794,176	RPGRIP1	snp	NM_020366.3	NP_065099.3	+	2,554	G	A	851	R	Q	MISSENSE	F01	1	2
chr14	22,102,841	OR10G2	snp	NM_001005466.1	NP_001005466.1	-	156	T	C	52	M	V	MISSENSE	D01	1	2
chr14	22,102,912	OR10G2	snp	NM_001005466.1	NP_001005466.1	-	85	C	T	28	S	N	MISSENSE	C01	2	2
chr14	23,299,145	MRPL52	snp	NM_180982.2	NP_851313.1	+	54	T	C	8	F	L	MISSENSE	F01	1	2
chr14	23,302,646	MRPL52	snp	NM_181304.2	NP_851821.1	+	390	A	G	-	M	V	MISSTART	D01	2	2
chr14	24,769,179	C14orf21	snp	NM_174913.1	NP_777573.1	+	82	C	T	6	S	F	MISSENSE	A01	2	2
chr14	24,769,808	C14orf21	snp	NM_174913.1	NP_777573.1	+	505	G	A	147	S	N	MISSENSE	B01	1	2
chr14	24,770,874	C14orf21	snp	NM_174913.1	NP_777573.1	+	817	C	G	251	T	S	MISSENSE	A01	2	2
chr14	24,773,417	C14orf21	snp	NM_174913.1	NP_777573.1	+	1,644	G	A	527	D	N	MISSENSE	A01	1	2
chr14	58,899,156	KIAA0586	del	NM_014749.3	NP_055564.3	+	664	G		130	R	K	FRAMESHIFT	D01	1	2

chr14	58,955,529	KIAA0586	snp	NM_014749.3	NP_055564.3	+	3,763	T	C	1,163	L	S	MISSENSE	F01	1	2
chr14	64,443,403	SYNE2	snp	NM_015180.4	NP_055995.4	+	1,481	C	G	417	Q	E	MISSENSE	B01	2	2
chr14	64,610,627	SYNE2	snp	NM_015180.4	NP_055995.4	+	15,674	C	T	5,148	R	C	MISSENSE	A01	2	2
chr14	104,642,758	KIF26A	snp	NM_015656.1	NP_056471.1	+	3,633	C	T	1,211	R	W	MISSENSE	B01	2	2
chr14	104,642,951	KIF26A	snp	NM_015656.1	NP_056471.1	+	3,826	T	C	1,275	M	T	MISSENSE	A01	1	2
chr14	105,415,406	AHNAK2	del	NM_138420.2	NP_612429.2	-	6,500	T		2,126	RA	RP	FRAMESHIFT	B01	1	3
chr14	105,418,302	AHNAK2	snp	NM_138420.2	NP_612429.2	-	3,604	C	T	1,161	R	K	MISSENSE	C01	2	3
chr14	105,419,134	AHNAK2	snp	NM_138420.2	NP_612429.2	-	2,772	G	A	884	P	S	MISSENSE	A01	1	3
chr15	34,649,536	C15orf55	snp	NM_175741.1	NP_786883.1	+	3,398	G	A	1,081	A	T	MISSENSE	E01	1	2
chr15	34,649,615	C15orf55	snp	NM_175741.1	NP_786883.1	+	3,477	G	A	1,107	R	Q	MISSENSE	B01	1	2
chr15	42,120,341	JMJD7-PLA2G4B	snp	NM_005090.3	NP_005081.1	+	59	A	G	6	E	G	MISSENSE	C01	2	2
chr15	42,120,341	JMJD7-PLA2G4B	snp	NM_005090.3	NP_005081.1	+	59	A	G	6	E	G	MISSENSE	F01	2	2
chr15	42,120,341	JMJD7	snp	NM_001114632.1	NP_001108104.1	+	59	A	G	6	E	G	MISSENSE	C01	2	2
chr15	42,120,341	JMJD7	snp	NM_001114632.1	NP_001108104.1	+	59	A	G	6	E	G	MISSENSE	F01	2	2
chr1	42,974,699	STARD9	sub	NM_020759.2	NP_065810.2	+	2,520	A	CT	821	R	L	FRAMESHIFT	D01	2	4

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chr15	42,974,703	STARD9	del	NM_020759.2	NP_065810.2	+	2,524	G		822	S	T	FRAMESHIFT	D01	2	4
chr15	42,985,989	STARD9	snp	NM_020759.2	NP_065810.2	+	12,270	A	G	4,071	T	A	MISSENSE	B01	2	4
chr15	43,008,922	STARD9	snp	NM_020759.2	NP_065810.2	+	13,389	C	T	4,444	R	*	NONSENSE	E01	2	4
chr15	43,011,689	STARD9	snp	NM_020759.2	NP_065810.2	+	14,077	G	A	4,673	G	D	MISSENSE	A01	1	4
chr15	63,046,034	TLN2	snp	NM_015059.2	NP_055874.2	+	4,395	C	T	1,465	P	S	MISSENSE	A01	2	2
chr15	63,097,808	TLN2	snp	NM_015059.2	NP_055874.2	+	6,487	C	T	2,162	S	L	MISSENSE	B01	2	2
chr15	72,190,293	MYO9A	snp	NM_006901.2	NP_008832.2	-	5,022	C	T	1,516	R	H	MISSENSE	C01	2	2
chr15	72,190,695	MYO9A	snp	NM_006901.2	NP_008832.2	-	4,620	G	A	1,382	A	V	MISSENSE	B01	1	2
chr15	76,673,927	SCAPER	snp	NM_001145923.1	NP_001139395.1	-	3,697	G	C	919	Q	E	MISSENSE	C01	1	2
chr15	76,696,900	SCAPER	snp	NM_001145923.1	NP_001139395.1	-	3,632	T	C	897	H	R	MISSENSE	A01	1	2
chr16	812,699	MSLN	snp	NM_005823.5	NP_005814.2	+	187	G	A	6	R	Q	MISSENSE	B01	1	2
chr16	818,521	MSLN	del	NM_005823.5	NP_005814.2	+	1,907	CG		579	GG	GH	FRAMESHIFT	E01	2	2
chr16	4,625,930	LOC342346	snp	NM_001145011.1	NP_001138483.1	+	1,449	C	T	483	R	C	MISSENSE	D01	2	2
chr16	4,650,313	LOC342346	snp	NM_001145011.1	NP_001138483.1	+	3,421	C	T	1,140	P	L	MISSENSE	A01	2	2

chr16	4,934,634	PPL	snp	NM_002705.4	NP_002696.3	-	4,110	C	T	1,340	E	K	MISSENSE	D01	2	2
chr16	4,935,911	PPL	snp	NM_002705.4	NP_002696.3	-	2,833	C	A	914	S	I	MISSENSE	B01	1	2
chr16	10,775,948	TEKT5	snp	NM_144674.1	NP_653275.1	-	835	T	C	254	D	G	MISSENSE	B01	1	2
chr16	10,788,293	TEKT5	snp	NM_144674.1	NP_653275.1	-	508	G	A	145	S	L	MISSENSE	D01	1	2
chr16	20,944,599	DNAH3	snp	NM_017539.1	NP_060009.1	-	12,226	C	T	4,075	R	K	MISSENSE	A01	2	2
chr16	21,049,219	DNAH3	snp	NM_017539.1	NP_060009.1	-	4,812	C	T	1,604	A	T	MISSENSE	C01	2	2
chr16	28,403,354	EIF3CL	del	NM_001099661.1	NP_001093131.1	-	971	CTC		294	E		DELETE	D01	1	2
chr16	28,403,363	EIF3CL	del	NM_001099661.1	NP_001093131.1	-	962	CTC		294	E		DELETE	B01	1	2
chr16	70,897,086	HYDIN	snp	NM_032821.2	NP_116210.2	-	11,595	T	C	3,822		CTGATCGATTTGCGG	DISRUPT	A01	1	3
chr16	70,928,376	HYDIN	snp	NM_032821.2	NP_116210.2	-	9,347	G	A	3,073	R	C	MISSENSE	E01	1	3
chr16	70,952,280	HYDIN	snp	NM_032821.2	NP_116210.2	-	7,961	C	T	2,611	A	T	MISSENSE	C01	2	3
chr16	88,494,602	ZNF469	sub	NM_001127464.1	NP_001120936.1	+	724	GC	TT	241	S	I	MISSENSE	C01	2	2
chr16	88,495,574	ZNF469	snp	NM_001127464.1	NP_001120936.1	+	1,696	C	T	565	A	V	MISSENSE	D01	1	2
chr16	88,804,363	FAM38A	snp	NM_001142864.2	NP_001136336.2	-	997	C	T	332	R	H	MISSENSE	F01	2	2
chr1	88,808,727	FAM38A	snp	NM_001142864.2	NP_001136336.2	-	262	T	C	87	D	G	MISSENSE	A01	1	2

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chr17	650,206	GEMIN4	snp	NM_015721.2	NP_056536.2	-	1,194	G	A	358	A	V	MISSENSE	B01	1	2
chr17	650,795	GEMIN4	snp	NM_015721.2	NP_056536.2	-	605	C	T	162	V	I	MISSENSE	F01	2	2
chr17	11,501,864	DNAH9	snp	NM_001372.3	NP_001363.2	+	117	G	T	16	G	V	MISSENSE	C01	2	2
chr17	11,659,960	DNAH9	snp	NM_001372.3	NP_001363.2	+	6,882	G	A	2,271	R	H	MISSENSE	A01	1	2
chr17	17,698,855	RAI1	snp	NM_030665.3	NP_109590.3	+	3,062	T	C	864	L	P	MISSENSE	C01	1	2
chr17	17,700,042	RAI1	del	NM_030665.3	NP_109590.3	+	4,249	GAG		1,260	E		DELETE	F01	1	2
chr17	18,881,468	FAM83G	snp	NM_001039999.2	NP_001035088.2	-	1,676	C	A	503	V	L	MISSENSE	A01	1	2
chr17	18,881,604	FAM83G	snp	NM_001039999.2	NP_001035088.2	-	1,540	C	A	457	Q	H	MISSENSE	E01	1	2
chr17	33,680,507	SLFN11	snp	NM_001104590.1	NP_001098060.1	-	2,122	C	T	589	R	H	MISSENSE	D01	2	3
chr17	33,690,314	SLFN11	snp	NM_001104590.1	NP_001098060.1	-	865	T	C	170	K	R	MISSENSE	B01	1	3
chr17	33,690,425	SLFN11	snp	NM_001104590.1	NP_001098060.1	-	754	C	T	133	R	H	MISSENSE	A01	2	3
chr17	33,806,513	LOC100506736	snp	XM_003118755.1	XP_003118803.1	-	1,680	T	A	267	I	F	MISSENSE	F01	1	2
chr17	33,807,028	LOC100506736	sub	XM_003118755.1	XP_003118803.1	-	1,164	CA	GAG	95	CA	LC	FRAMESHIFT	E01	2	2
chr17	34,186,055	C17orf66	snp	NM_152781.2	NP_689994.2	-	923	C	T	258	E	K	MISSENSE	E01	2	2

chr17	34,192,350	C17orf66	snp	NM_152781.2	NP_689994.2	-	336	G	A	62	P	L	MISSENSE	C01	1	2
chr17	35,896,177	SYNRG	snp	NM_198882.1	NP_942583.1	-	3,417	G	A	1,111	A	V	MISSENSE	D01	1	2
chr17	35,902,203	SYNRG	snp	NM_198882.1	NP_942583.1	-	2,920	A	C	945	F	L	MISSENSE	B01	2	2
chr17	36,623,094	ARHGAP23	snp	NM_020876.1	NP_065927.1	+	1,550	G	T	296	A	S	MISSENSE	D01	2	2
chr17	36,623,094	ARHGAP23	snp	NM_020876.1	NP_065927.1	+	1,550	G	T	296	A	S	MISSENSE	F01	2	2
chr17	39,183,016	KRTAP1-5	del	NM_031957.1	NP_114163.1	-	437	G		131	PC	PA	FRAMESHIFT	B01	2	2
chr17	39,183,016	KRTAP1-5	del	NM_031957.1	NP_114163.1	-	437	G		131	PC	PA	FRAMESHIFT	C01	1	2
chr17	39,254,246	KRTAP4-8	snp	NM_031960.2	NP_114166.1	-	128	A	T	29	C	*	NONSENSE	D01	2	2
chr17	39,254,246	KRTAP4-8	snp	NM_031960.2	NP_114166.1	-	128	A	T	29	C	*	NONSENSE	F01	2	2
chr17	39,254,256	KRTAP4-8	snp	NM_031960.2	NP_114166.1	-	118	G	T	26	P	H	MISSENSE	D01	2	2
chr17	39,254,256	KRTAP4-8	snp	NM_031960.2	NP_114166.1	-	118	G	T	26	P	H	MISSENSE	F01	2	2
chr17	55,183,854	AKAP1	ins	NM_003488.3	NP_003479.1	+	1,260		GGCTTGGATAGAAATGAGGAG	343		GLDRNEE	INSERT	A01	1	2
chr17	55,183,854	AKAP1	ins	NM_003488.3	NP_003479.1	+	1,260		GGCTTGGATAGAAATGAGGAG	343		GLDRNEE	INSERT	B01	1	2
chr17	56,329,651	LPO	snp	NM_006151.2	NP_006142.1	+	1,205	T	A	296	I	N	MISSENSE	E01	1	2
chr1	56,343,533	LPO	snp	NM_006151.2	NP_006142.1	+	1,855	C	T	513	R	W	MISSENSE	D01	2	2

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chr17	62,019,040	SCN4A	snp	NM_000334.4	NP_000325.4	-	4,677	G	A	1,533	T	M	MISSENSE	F01	1	2
chr17	62,022,043	SCN4A	ins	NM_000334.4	NP_000325.4	-	3,978		TCT	1,304		K	INSERT	C01	2	2
chr18	28,581,653	DSC3	snp	NM_024423.2	NP_077741.2	-	2,319	C	T	721	R	H	MISSENSE	A01	2	2
chr18	28,602,410	DSC3	snp	NM_024423.2	NP_077741.2	-	987	G	A	277	T	M	MISSENSE	C01	1	2
chr19	619,238	POLRMT	snp	NM_005035.3	NP_005026.3	-	3,179	C	T	1,041	E	K	MISSENSE	C01	2	2
chr19	632,848	POLRMT	snp	NM_005035.3	NP_005026.3	-	233	C	T	59	V	M	MISSENSE	F01	2	2
chr19	2,403,122	TMPRSS9	snp	NM_182973.1	NP_892018.1	+	497	T	A	165	C	*	NONSENSE	B01	1	2
chr19	2,421,963	TMPRSS9	snp	NM_182973.1	NP_892018.1	+	2,164	C	A	721	T	K	MISSENSE	D01	1	2
chr19	4,511,223	PLIN4	sub	NM_001080400.1	NP_001073869.1	-	2,703	GCC	ACT	901	G	S	MISSENSE	C01	1	3
chr19	4,511,749	PLIN4	snp	NM_001080400.1	NP_001073869.1	-	2,179	G	A	726	A	V	MISSENSE	F01	1	3
chr19	4,512,777	PLIN4	sub	NM_001080400.1	NP_001073869.1	-	1,150	AC	G	383	GT	AR	FRAMESHIFT	A01	1	3
chr19	4,512,780	PLIN4	snp	NM_001080400.1	NP_001073869.1	-	1,148	C	T	382	M	I	MISSENSE	A01	2	3
chr19	4,512,782	PLIN4	sub	NM_001080400.1	NP_001073869.1	-	1,140	TGACCAT	GTCAGGAC	380	MVM	VLT	FRAMESHIFT	A01	1	3
chr19	4,512,788	PLIN4	snp	NM_001080400.1	NP_001073869.1	-	1,140	T	C	380	M	V	MISSENSE	A01	2	3

chr19	6,755,178	SH2D3A	snp	NM_005490.2	NP_005481.2	-	837	C	T	214	R	Q	MISSENSE	A01	2	2
chr19	6,760,706	SH2D3A	snp	NM_005490.2	NP_005481.2	-	554	G	A	120	R	*	NONSENSE	F01	1	2
chr19	8,182,160	FBN3	ins	NM_032447.3	NP_115823.3	-	3,499		T	1,159	R	Q	FRAMESHIFT	A01	2	2
chr19	8,212,234	FBN3	snp	NM_032447.3	NP_115823.3	-	150	G	A	43	R	C	MISSENSE	B01	1	2
chr19	9,011,443	MUC16	snp	NM_024690.2	NP_078966.2	-	38,992	C	T	12,929	R	H	MISSENSE	E01	2	4
chr19	9,058,488	MUC16	snp	NM_024690.2	NP_078966.2	-	29,160	T	C	9,652	I	V	MISSENSE	A01	1	4
chr19	9,068,108	MUC16	snp	NM_024690.2	NP_078966.2	-	19,540	G	A	6,445	A	V	MISSENSE	A01	1	4
chr19	9,086,982	MUC16	snp	NM_024690.2	NP_078966.2	-	5,035	G	T	1,610	T	K	MISSENSE	F01	2	4
chr19	9,090,040	MUC16	snp	NM_024690.2	NP_078966.2	-	1,977	C	T	591	G	R	MISSENSE	B01	2	4
chr19	9,577,373	ZNF560	snp	NM_152476.2	NP_689689.2	-	2,458	C	T	749	R	H	MISSENSE	C01	2	2
chr19	9,579,829	ZNF560	snp	NM_152476.2	NP_689689.2	-	772	G	A	187	A	V	MISSENSE	A01	1	2
chr19	16,855,891	NWD1	snp	NM_001007525.2	NP_001007526.2	+	1,503	G	C	8	G	R	MISSENSE	E01	1	2
chr19	16,860,095	NWD1	snp	NM_001007525.2	NP_001007526.2	+	1,716	T	C	79	C	R	MISSENSE	E01	2	2
chr19	16,870,168	NWD1	snp	NM_001007525.2	NP_001007526.2	+	2,976	C	T	499	R	C	MISSENSE	C01	2	2
chr1	17,836,791	MAP1S	snp	NM_018174.4	NP_060644.4	+	607	A	G	199	Q	R	MISSENSE	A01	2	2

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chr19	17,837,759	MAP1S	snp	NM_018174.4	NP_060644.4	+	1,575	C	T	522	R	W	MISSENSE	C01	2	2
chr19	38,834,225	CATSPERG	snp	NM_021185.4	NP_067008.3	+	417	G	T	110	E	*	NONSENSE	E01	2	2
chr19	38,853,156	CATSPERG	del	NM_021185.4	NP_067008.3	+	2,385	A		766	S	A	FRAMESHIFT	A01	2	2
chr19	39,399,921	LOC643669	del	XM_933621.5	XP_938714.3	-	875	TTC		293	K		DELETE	F01	1	2
chr19	39,401,291	LOC643669	snp	XM_933621.5	XP_938714.3	-	744	T	C	248	R	G	MISSENSE	C01	1	2
chr19	41,114,407	LTBP4	sub	NM_003573.2	NP_003564.2	+	1,414	GC	CT	471	R	P	MISSENSE	C01	1	2
chr19	41,128,565	LTBP4	snp	NM_003573.2	NP_003564.2	+	3,567	G	A	1,189	E	K	MISSENSE	A01	1	2
chr19	41,173,870	NUMBL	ins	NM_004756.3	NP_004747.1	-	1,500		TTGCTGCTGCTG	445		QQQQ	INSERT	F01	2	2
chr19	41,179,455	NUMBL	snp	NM_004756.3	NP_004747.1	-	996	C	A	276	A	S	MISSENSE	A01	1	2
chr19	42,510,093	GRIK5	snp	NM_002088.3	NP_002079.3	-	2,078	G	A	681	R	W	MISSENSE	B01	1	2
chr19	42,558,583	GRIK5	snp	NM_002088.3	NP_002079.3	-	978	A	G	314	V	A	MISSENSE	A01	2	2
chr19	46,914,462	CCDC8	snp	NM_032040.3	NP_114429.2	-	2,456	T	G	534	E	D	MISSENSE	F01	1	2
chr19	46,915,113	CCDC8	snp	NM_032040.3	NP_114429.2	-	1,805	A	C	317	N	K	MISSENSE	D01	2	2
chr19	48,047,728	ZNF541	snp	NM_001101419.1	NP_001094889.1	-	2,056	G	C	685	T	S	MISSENSE	D01	1	2

chr19	48,047,728	ZNF541	snp	NM_001101419.1	NP_001094889.1	-	2,056	G	C	685	T	S	MISSENSE	F01	1	2
chr19	50,393,042	IL4I1	snp	NM_152899.1	NP_690863.1	-	1,665	C	T	529	V	M	MISSENSE	C01	2	2
chr19	50,399,160	IL4I1	snp	NM_152899.1	NP_690863.1	-	240	G	A	54	R	W	MISSENSE	E01	1	2
chr19	54,723,032	LILRB3	ins	NM_006864.2	NP_006855.2	-	1,502		G	464	R	P	FRAMESHIFT	E01	2	2
chr19	54,725,155	LILRB3	sub	NM_006864.2	NP_006855.2	-	863	TG	CA	250	YN	YD	MISSENSE	C01	1	2
chr19	57,640,668	USP29	snp	NM_020903.2	NP_065954.1	+	981	A	T	208	N	I	MISSENSE	C01	2	2
chr19	57,642,393	USP29	snp	NM_020903.2	NP_065954.1	+	2,706	T	C	783	L	P	MISSENSE	E01	1	2
chr19	58,773,338	ZNF544	snp	NM_014480.2	NP_055295.2	+	1,840	G	T	455	R	I	MISSENSE	E01	2	2
chr19	58,773,540	ZNF544	snp	NM_014480.2	NP_055295.2	+	2,042	C	G	522	N	K	MISSENSE	A01	2	2
chr20	3,211,845	SLC4A11	snp	NM_032034.3	NP_114423.1	-	1,086	G	A	346	R	W	MISSENSE	D01	1	2
chr20	3,218,664	SLC4A11	snp	NM_001174090.1	NP_001167561.1	-	170	C	T	15	D	N	MISSENSE	E01	1	2
chr20	6,065,917	FERMT1	snp	NM_017671.4	NP_060141.3	-	2,176	T	C	462	Q	R	MISSENSE	A01	1	2
chr20	6,077,648	FERMT1	snp	NM_017671.4	NP_060141.3	-	1,777	T	A	329	E	V	MISSENSE	C01	1	2
chr20	30,330,341	TPX2	snp	NM_012112.4	NP_036244.2	+	521	A	T	-		TTCTGCTTCTTCCTG	DISRUPT	F01	2	2
chr2	30,370,111	TPX2	snp	NM_012112.4	NP_036244.2	+	1,812	T	A	371	L	Q	MISSENSE	A01	1	2

0																
chr20	32,664,916	RALY	snp	NM_007367.2	NP_031393.2	+	991	G	A	231	G	S	MISSENSE	E01	1	2
chr20	32,664,916	RALY	snp	NM_007367.2	NP_031393.2	+	991	G	A	231	G	S	MISSENSE	F01	1	2
chr20	39,978,435	LPIN3	snp	NM_022896.1	NP_075047.1	+	751	T	A	220	S	T	MISSENSE	D01	1	2
chr20	39,987,410	LPIN3	snp	NM_022896.1	NP_075047.1	+	2,551	C	T	820	R	C	MISSENSE	B01	2	2
chr20	46,262,805	NCOA3	snp	NM_181659.2	NP_858045.1	+	1,239	C	T	326	H	Y	MISSENSE	A01	2	2
chr20	46,264,405	NCOA3	snp	NM_181659.2	NP_858045.1	+	1,713	C	T	484	R	C	MISSENSE	B01	2	2
chr20	61,451,332	COL9A3	snp	NM_001853.3	NP_001844.3	+	310	G	A	102	R	Q	MISSENSE	B01	1	2
chr20	61,461,757	COL9A3	snp	NM_001853.3	NP_001844.3	+	1,210	G	C	402	G	A	MISSENSE	C01	1	2
chr20	61,470,064	COL9A3	snp	NM_001853.3	NP_001844.3	+	1,818	G	A	605	A	T	MISSENSE	B01	1	2
chr20	61,939,414	COL20A1	del	NM_020882.2	NP_065933.2	+	847	CGC		249	R		DELETE	F01	2	3
chr20	61,939,928	COL20A1	snp	NM_020882.2	NP_065933.2	+	910	C	G	270	Q	E	MISSENSE	B01	2	3
chr20	61,943,841	COL20A1	snp	NM_020882.2	NP_065933.2	+	1,973	G	A	624	R	H	MISSENSE	A01	1	3
chr21	38,568,210	TTC3	snp	NM_001001894.1	NP_001001894.1	+	5,557	G	T	1,817	R	L	MISSENSE	B01	2	2
chr21	38,568,251	TTC3	snp	NM_001001894.1	NP_001001894.1	+	5,598	C	T	1,831	R	C	MISSENSE	A01	2	2

chr21	43,274,689	PRDM15	snp	NM_001040424.1	NP_001035514.1	-	735	C	T	211	G	S	MISSENSE	C01	1	2
chr21	43,282,032	PRDM15	snp	NM_022115.3	NP_071398.3	-	812	C	T	233		ATGTGT	DISRUPT	A01	2	2
chr22	22,283,408	LOC100507442	snp	XM_003118824.1	XP_003118872.1	-	1,096	A	G	111	W	R	MISSENSE	D01	2	2
chr22	22,283,410	LOC100507442	snp	XM_003118824.1	XP_003118872.1	-	1,094	G	A	110	A	V	MISSENSE	C01	1	2
chr22	30,517,769	HORMAD2	snp	NM_152510.2	NP_689723.1	+	624	A	G	186	Y	C	MISSENSE	C01	2	2
chr22	30,518,165	HORMAD2	snp	NM_152510.2	NP_689723.1	+	846	G	A	260	G	D	MISSENSE	E01	1	2
chr22	30,776,105	RNF215	snp	NM_001017981.1	NP_001017981.1	-	952	G	C	317	P	R	MISSENSE	C01	1	2
chr22	30,776,310	RNF215	snp	NM_001017981.1	NP_001017981.1	-	823	C	T	274	R	Q	MISSENSE	B01	2	2
chr22	42,609,792	TCF20	snp	NM_181492.1	NP_852469.1	-	1,652	G	C	506	P	A	MISSENSE	E01	1	2
chr22	42,610,036	TCF20	ins	NM_181492.1	NP_852469.1	-	1,409		G	426	S	Q	FRAMESHIFT	E01	2	2
chr22	42,610,594	TCF20	ins	NM_181492.1	NP_852469.1	-	851		GGA	246		S	INSERT	F01	2	2
chr22	50,278,661	ZBED4	ins	NM_014838.2	NP_055653.2	+	1,821		AAAAGGTCATG	455	RLKSE	RS*	NONSENSE	A01	2	2
chr22	50,278,661	ZBED4	ins	NM_014838.2	NP_055653.2	+	1,821		AAAAGGTCATG	455	RLKSE	RS*	NONSENSE	C01	2	2
chrX	9,935,685	LOC100288814	snp	NM_001195081.1	NP_001182010.1	+	288	A	G	96	S	G	MISSENSE	A01	2	2
chrX	9,935,850	LOC100288814	snp	NM_001195081.1	NP_001182010.1	+	453	G	A	151	V	I	MISSENSE	E01	2	2
ch	66,765,26	AR	ins	NM_000044.2	NP_000035.2	+	1,38		CAG	91		Q	INSERT	B0	1	2

rX	1						8							1		
chrX	66,941,750	AR	snp	NM_000044.2	NP_000035.2	+	3,509	C	G	798	Q	E	MISSENSE	E01	2	2
chrX	70,516,495	NONO	ins	NM_007363.4	NP_031389.3	+	1,327		A	243	NQ	KP	FRAMESHIFT	C01	1	2
chrX	70,518,594	NONO	snp	NM_007363.4	NP_031389.3	+	1,805	G	A	403	A	T	MISSENSE	E01	1	2
chrX	84,601,034	POF1B	snp	NM_024921.3	NP_079197.3	-	699	T	C	184	Q	R	MISSENSE	A01	1	2
chrX	84,634,413	POF1B	ins	NM_024921.3	NP_079197.3	-	192		AGCTGCTGGG	20	EVLQ	AAPR	FRAMESHIFT	D01	1	2
chrX	152,584,958	LOC649201	snp	XM_001127211.3	XP_001127211.3	-	1,314	G	A	438	R	C	MISSENSE	A01	1	3
chrX	152,586,141	LOC649201	snp	XM_001127211.3	XP_001127211.3	-	572	T	C	190	I	M	MISSENSE	C01	1	3
chrX	152,587,001	LOC649201	snp	XM_001127211.3	XP_001127211.3	-	112	C	T	37	R	K	MISSENSE	F01	1	3

Genes for which novel deviations from reference genome were identified in two or more of the six index SMZL genomes are shown. Alterations affecting the coding region or splicesome recognition sequence of the indicated genes not present in dbSNP are listed in chromosomal order. Var: The type of deviation from reference sequence identified; snp- single nucleotide polymorphism, ins- insertion, del- deletion, sub- substitution. Str: strandedness of the gene. Ref: reference sequence nucleotide or amino acid at the position indicated. Call: called sequence for of the indicated SMZL genome. Impact- predicted functional consequence on protein coding of deviation indicated. bp- base pair, aa- amino acid. Gen: genome identifier, corresponds to Figure 1 legend. A01, B01, C01 are the *NOTCH2* mutated genomes depicted in Figure 2. All: Allele. No: number of genomes with sequence deviation from reference (of six total). Asterisk indicates stop codon. Mutations identified in *MAML2* by whole genome sequencing or by direct Sanger DNA sequencing are also shown.

Table S3. Primers used for amplification prior to Sanger sequencing.

					Amino Acid Residues	
Fragment	**Forward Primer Sequence for Amplification**	**Reverse Primer Sequence for Amplification**	**Size (nt)**	**Exon**	**Begin**	**End**
34	GTGGAGGTTTTCTAGAAACCTCA	GCACAATACTGGCTCAGACAG	371	25	1335	1420
35	GAGTCAGGCTGTGCCAGTA	CTGTTGCAGGCCTCATCACA	212	25	1385	1430
36	GGTAGCCGCTGTGAACTCTA	CGAGAAACTGAAGTGTGTTAGTGA	351	25	1420	1504
37	AGCTCCAGTCTAATCTGAGCTCT	CAGGTGGCATCAATACCACA	208	26	1505	1545
38	GGTGCAACAGTGAGGAGTGT	TAGCCTTGAAGTTCAGAAACCA	329	26	1530	1620
39	ATGACATGTTCTGCCTGACCT	CCTTTACACCAGTGCCACTC	244	27	1621	1668
40	ATCTAATGCTGACATTGAGAGGT	AGAGAGAGCCATGCTTACGCT	192	28	1669	1700
41	GTTGCTGTTGTCATCATTCTGT	AATCATGATTCAACAAGATATGC	244	28	1690	1738
42	GTGTCATGGTGGAAAGTGTTG	CAGATAATGGCTGACAATGGTG	221	29	1739	1770
43	AAACAATGGGAGATAAGCAGCGGTGGTG	GACAACAATGTGGAACCATG	337	30	1771	1827
44	CAAATAGAGCTGTTTCAACCATAG	ATTGGCATCTGCACCTGCATC	310	31	1828	1895
45	AGATGCAGAGGACTCTTCTGCT	TATTATTCAAGTGACTCTTCTCATGTT	268	31	1870	1927
46	CTACACTGTAGCCTCAGCTCTGAT	CCAAATCCCTGCCTTTCATC	274	32	1928	1977
47	CATTGTGCAAGTCATAGTGTCTT	GAATGGGCTTATAACTGAGGCA	233	33	1977	2009
48	CTCAAGAGTGTTATTAACATGTGTTC	CTTCAGGCTGAGGAAAGATCTG	305	34	2010	2080
49	TCGCATGCACCATGACATTG	GGATAAAGTTACTGAACTCTCAGAC	292	34	2060	2140
50	TTGCCAAGGAGGCAAAGGATG	CTCACTGAGGGAAGCACAGT	310	34	2130	2210
51	TGGGATCTTACAGGCCTCAC	CCAGGACCATACCAAACATC	299	34	2185	2260
52	CGCATGGAGGTGAATGAGA	CCATTTCTGGAATCTGGTACAT	271	34	2260	2335
53	CTAAAGGCAGTATTGCCCAAC	CTGGAGGTGACCACTGTGAC	296	34	2320	2400
54	GGCAGGTAGCTCAGACCAT	GTCAGGAGACTCTGGGGAT	182	34	2365	2415
55	GCTGAGCGAACACCCAGT	TGTTCCTCAGCAGCATTTACA	283	34	2410	2471

Primer sequences for PCR amplification of *NOTCH2* gene C-terminus exon regions are shown. PCR primers were designed to amplify 200-300 base-pair products within *NOTCH2* coding regions with sufficient overlapping coverage between adjacent fragments to ensure reliable base-calling. For sequencing reactions, nested primer sequences offset by 3-4 nucleotides from the corresponding amplification primer were used to improve sequencing specificity. Product sizes as well as exon and amino acid residue coverage are also listed.

Table S4. Nested primers used for Sanger sequencing.

Fragment	Forward Primer Sequence for Sequencing	Reverse Primer Sequence for Sequencing
34	AGGTTTTCTAGAAACCTCAAACT	AATACTGGCTCAGACAGGTGG
35	CAGGCTGTGCCAGTAGCCC	TGCAGGCCTCATCACAGACG
36	GCCGCTGTGAACTCTACACG	AAACTGAAGTGTGTTAGTGACAGT
37	CCAGTCTAATCTGAGCTCTTTTG	TGGCATCAATACCACAATAA
38	CAACAGTGAGGAGTGTGGTT	CTTGAAGTTCAGAAACCAAACA
39	CATGTTCTGCCTGACCTGCAC	CCTTTACACCAGTGCCACTC
40	AATGCTGACATTGAGAGGTTAAT	AGAGCCATGCTTACGCTTTCG
41	CTGTTGTCATCATTCTGTTTAT	ATGATTCAACAAGATATGCTTTT
42	CATGGTGGAAAGTGTTGAAAA	TAATGGCTGACAATGGTGGTTC
43	AATGGGAGATAAGCAGCGGTGGTGGAGGCTC	ACAATGTGGAACCATGGGCA
44	TAGAGCTGTTTCAACCATAGGGTT	GCATCTGCACCTGCATCCAGG
45	GCAGAGGACTCTTCTGCTAACA	ATTCAAGTGACTCTTCTCATGTTCTTTACC
46	ACTGTAGCCTCAGCTCTGATGCCC	ATCCCTGCCTTTCATCCCTA
47	GTGCAAGTCATAGTGTCTTATAC	GGGCTTATAACTGAGGCACTGC
48	AGAGTGTTATTAACATGTGTTCTGTG	AGGCTGAGGAAAGATCTGTTGG
49	ATGCACCATGACATTGTGCG	AAAGTTACTGAACTCTCAGACAGTT
50	CAAGGAGGCAAAGGATGCCAA	CTGAGGGAAGCACAGTGCTG
51	ATCTTACAGGCCTCACCCAA	GACCATACCAAACATCTCAT
52	ATGGAGGTGAATGAGACCC	CCATTTCTGGAATCTGGTACAT
53	AGGCAGTATTGCCCAACCAGC	AGGTGACCACTGTGACTGGG
54	GGTAGCTCAGACCATTCTC	GGAGACTCTGGGGATGGTG
55	AGCGAACACCCAGTCACA	CCTCAGCAGCATTTACAAAAG

Primer sequences for PCR amplification of *NOTCH2* gene C-terminus exon regions are shown. PCR primers were designed to amplify 200-300 base-pair products within *NOTCH2* coding regions with sufficient overlapping coverage between adjacent fragments to ensure reliable base-calling. For sequencing reactions, nested primer sequences offset by 3-4 nucleotides from the corresponding amplification primer were used to improve sequencing specificity. Product sizes as well as exon and amino acid residue coverage are als