HGVSg SNV	HGVSc	HGVSp	Consequence	Gene SYMBOL	Ensembl Gene	Ensembl transcript	MANE_SELECT
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NC_000001.11:g.8359847G>A	ENST00000337907.7:c.3535C>T	ENSP00000338629.3:p.Arg1179Te	stop_gained	RERE	ENSG00000142599	ENST00000337907.7	
NC_000001.11:g.9972126A>G	ENST00000377205.6:c.53A>G	ENSP00000366410.1:p.Asn18Ser	missense_variant	NMNAT1	ENSG00000173614	ENST00000377205.6	NM_022787.4
NC_000001.11:g.11130747C>T	ENST00000361445.9:c.5395G>A	ENSP00000354558.4:p.Glu1799Ly	missense_variant	MTOR	ENSG00000198793	ENST00000361445.9	NM_004958.4
NC_000001.11:g.11533815_11533818dup	ENST00000294484.7:c.3376-566_3376-563dup		intron_variant	DISP3	ENSG00000204624	ENST00000294484.7	NM_020780.2
NC_000001.11:g.12276238dup			upstream_gene_variant	VPS13D	ENSG00000048707	ENST00000011700.10	
NC_000001.11:g.12456014C>T	ENST00000011700.10:c.8816C>T	ENSP00000011700.6:p.Ser2939Le	missense_variant	VPS13D	ENSG00000048707	ENST00000011700.10	
NC_000001.11:g.15445606G>A	ENST00000375943.6:c.*103G>A		3_prime_UTR_variant	CTRC	ENSG00000162438	ENST00000375943.6	
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NC_000001.11:g.23796234C>A	ENST00000374497.7:c.905G>T	ENSP00000363621.3:p.Gly302Val	missense_variant	GALE	ENSG00000117308	ENST00000374497.7	
NC_000001.11:g.25811490del	ENST00000354177.9:c.876del	ENSP00000346109.5:p.Ser293Val	frameshift_variant	SELENON	ENSG00000162430	ENST00000354177.9	
NC_000001.11:g.27548578dup	ENST00000247087.10:c.3543dup	ENSP00000247087.4:p.Phe1182V	frameshift_variant	AHDC1	ENSG00000126705	ENST00000247087.10	
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NC_000001.11:g.35901541T>C	ENST00000373204.6:c.1088T>C	ENSP00000362300.4:p.Ile363Thr	missense_variant	AGO1	ENSG00000092847	ENST00000373204.6	NM_012199.5
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NC_000001.11:g.42456971C>G	ENST00000372556.3:c.64-276C>G		intron_variant	PPCS	ENSG00000127125	ENST00000372556.3	
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NC_000001.11:g.43431455A>G			downstream_gene_variant	SZT2	ENSG00000198198	ENST00000470139.1	
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NC_000001.11:g.45327284G>T	ENST00000334815.6:c.136G>T	ENSP00000335060.3:p.Ala46Ser	missense_variant	HPDL	ENSG00000186603	ENST00000334815.6	NM_032756.4
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NC_000001.11:g.45342466G>A			upstream_gene_variant	MUTYH	ENSG00000132781	ENST00000354383.10	
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NC_000001.11:g.68438951C>T	ENST00000262340.6:c.989G>A	ENSP00000262340.5:p.Cys330Tyr	missense_variant	RPE65	ENSG00000116745	ENST00000262340.6	NM_000329.3
NC_000001.11:g.68439023G>A	ENST00000262340.6:c.917C>T	ENSP00000262340.5:p.Thr306Ile	missense_variant	RPE65	ENSG00000116745	ENST00000262340.6	NM_000329.3
NC_000001.11:g.68440977dup	ENST00000262340.6:c.519dup	ENSP00000262340.5:p.Val174Cys	frameshift_variant	RPE65	ENSG00000116745	ENST00000262340.6	NM_000329.3
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NC_000001.11:g.109628807G>A	ENST00000256578.8:c.1571+1G>A		splice_donor_variant	AMPD2	ENSG00000116337	ENST00000256578.8	
NC_000001.11:g.113093504G>C	ENST00000361127.6:c.455G>C	ENSP00000355396.4:p.Ser152Thr	missense_variant	LRIG2	ENSG00000198799	ENST00000361127.6	NM_014813.3
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NC_000001.11:g.119076592G>A	ENST00000235521.5:c.106C>T	ENSP00000235521.4:p.Arg36Ter	stop_gained	WARS2	ENSG00000116874	ENST00000235521.5	NM_015836.4
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NC_000001.11:g.119997222_119997223del	ENST00000256646.7:c.525_526del	ENSP00000256646.2:p.Gly176Alaf	frameshift_variant	NOTCH2	ENSG00000134250	ENST00000256646.7	NM_024408.4
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NC_000001.11:g.151411741C>A	ENST00000271715.7:c.1810G>T	ENSP00000271715.2:p.Glu604Ter	stop_gained	POGZ	ENSG00000143442	ENST00000271715.7	NM_015100.4
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NC_000001.11:g.152312605_152312608del	ENST00000368799.2:c.2282_2285del	ENSP00000357789.1:p.Ser761Cys	frameshift_variant	FLG	ENSG00000143631	ENST00000368799.2	NM_002016.2
NC_000001.11:g.154589399G>A	ENST00000368471.8:c.1847C>T	ENSP00000357456.3:p.Ala616Val	missense_variant	ADAR	ENSG00000160710	ENST00000368471.8	
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NC_000001.11:g.160041881G>A	ENST00000509700.2:c.625C>T	ENSP00000491416.1:p.Leu209Ph	missense_variant	KCNJ10	ENSG00000177807	ENST00000509700.2	

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NC_000001.11:g.197434829T>C	ENST00000367397.1:c.1109T>C	ENSP00000356367.1:p.Ile370Thr	missense_variant	CRB1	ENSG00000134376	ENST00000367397.1	
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NC_000001.11:g.216422208del	ENST00000307340.8:c.131del	ENSP00000305941.3:p.Gly44Glufs	frameshift_variant	USH2A	ENSG00000042781	ENST00000307340.8	NM_206933.4
NC_000001.11:g.220114129T>G	ENST00000366922.3:c.1480-185T>G		intron_variant	IARS2	ENSG00000067704	ENST00000366922.3	NM_018060.4
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NC_000001.11:g.225404656G>A	ENST00000272163.9:c.1534C>T	ENSP00000272163.4:p.Arg512Trp	missense_variant	LBR	ENSG00000143815	ENST00000272163.9	NM_002296.4
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NC_000001.11:g.226071433C>G	ENST00000366813.1:c.365C>G	ENSP00000355778.1:p.Pro122Arg	missense_variant	H3-3A	ENSG00000163041	ENST00000366813.1	
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NC_000002.12:g.37147715G>C	ENST00000233057.9:c.92C>G	ENSP00000233057.4:p.Pro31Arg	missense_variant	EIF2AK2	ENSG00000055332	ENST00000233057.9	NM_001135651.3
NC_000002.12:g.45976483G>C	ENST00000306156.8:c.467G>C	ENSP00000306124.3:p.Arg156Pro	missense_variant	PRKCE	ENSG00000171132	ENST00000306156.8	NM_005400.3
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NC_000002.12:g.207088525C>T	ENST00000309446.11:c.790G>A	ENSP00000309570.6:p.Asp264Asn	missense_variant	KLF7	ENSG00000118263	ENST00000309446.11	NM_003709.4
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NC_000002.12:g.240783105C>T	ENST00000320389.12:c.803G>A	ENSP00000322791.8:p.Gly268Glu	missense_variant	KIF1A	ENSG00000130294	ENST00000320389.12	
NC_000003.12:g.4693644C>T	ENST00000302640.13:c.4157C>T	ENSP00000306253.9:p.Thr1386M	missense_variant&NMD_tra	ITPR1	ENSG00000150995	ENST00000302640.13	
NC_000003.12:g.4702871C>G	ENST00000302640.13:c.4551C>G	ENSP00000306253.9:p.Phe1517L	missense_variant&NMD_tra	ITPR1	ENSG00000150995	ENST00000302640.13	
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NC_000003.12:g.9742148_9742151del	ENST00000383829.7:c.1978_1981del	ENSP00000373340.2:p.Ser660Arg	frameshift_variant	BRPF1	ENSG00000156983	ENST00000383829.7	NM_001003694.2
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NC_000003.12:g.11031181G>A	ENST00000287766.10:c.1328G>A	ENSP00000287766.4:p.Gly443Asp	missense_variant	SLC6A1	ENSG00000157103	ENST00000287766.10	NM_003042.4
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NC_000003.12:g.30691477C>T	ENST00000295754.10:c.1582C>T	ENSP00000295754.5:p.Arg528Cys	missense_variant	TGFBR2	ENSG00000163513	ENST00000295754.10	NM_003242.6
NC_000003.12:g.32997049A>C	ENST00000307363.10:c.2030T>G	ENSP00000306920.4:p.Val677Gly	missense_variant	GLB1	ENSG00000170266	ENST00000307363.10	NM_000404.4
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NC_000003.12:g.33070576G>C	ENST00000307363.10:c.246-1606C>G		intron_variant	GLB1	ENSG00000170266	ENST00000307363.10	NM_000404.4
NC_000003.12:g.33134245_33134246del	ENST00000320954.11:c.1132_1133del	ENSP00000323696.5:p.Asn378Tyr	frameshift_variant	CRTAP	ENSG00000170275	ENST00000320954.11	NM_006371.5
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NC_000003.12:g.45496375T>C	ENST00000265537.8:c.1622+2T>C		splice_donor_variant&NMD_	LARS2	ENSG00000011376	ENST00000265537.8	
NC_000003.12:g.45513187C>T	ENST00000265537.8:c.*203C>T		3_prime_UTR_variant&NMI	LARS2	ENSG00000011376	ENST00000265537.8	
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NC_000003.12:g.58134734C>T	ENST00000295956.9:c.4633C>T	ENSP00000295956.5:p.Arg1545Te	stop_gained	FLNB	ENSG00000136068	ENST00000295956.9	NM_001457.4
NC_000003.12:g.58143497A>G	ENST00000295956.9:c.5309A>G	ENSP00000295956.5:p.Lys1770Ar	missense_variant	FLNB	ENSG00000136068	ENST00000295956.9	NM_001457.4
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NC_000003.12:g.70970835T>C	ENST00000318789.11:c.1653-30A>G		intron_variant	FOXP1	ENSG00000114861	ENST00000318789.11	
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NC_000003.12:g.87273501T>A	ENST00000344265.8:c.143-5A>T		splice_region_variant&splice	POU1F1	ENSG00000064835	ENST00000344265.8	
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NC_000003.12:g.119947348G>A	ENST00000264235.13:c.286C>T	ENSP00000264235.9:p.Arg96Ter	stop_gained	GSK3B	ENSG00000082701	ENST00000264235.13	NM_001146156.2
NC_000003.12:g.123352464G>A	ENST00000309879.9:c.202C>T	ENSP00000308685.5:p.Arg68Trp	missense_variant	ADCY5	ENSG00000173175	ENST00000309879.9	
NC_000003.12:g.123447695A>G	ENST00000462833.6:c.851T>C	ENSP00000419361.1:p.Met284Thr	missense_variant	ADCY5	ENSG00000173175	ENST00000462833.6	NM_183357.3
NC_000003.12:g.125232874G>A	ENST00000360647.9:c.1852C>T	ENSP00000353863.4:p.Gln618Ter	stop_gained	ZNF148	ENSG00000163848	ENST00000360647.9	NM_021964.3
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NC_000003.12:g.129488397G>A	ENST00000296266.7:c.2145G>A	ENSP00000296266.3:p.Lys715%3	splice_region_variant&syno	IFT122	ENSG00000163913	ENST00000296266.7	
NC_000003.12:g.129530914G>T	ENST00000296271.4:c.400G>T	ENSP00000296271.3:p.Glu134Ter	stop_gained	RHO	ENSG00000163914	ENST00000296271.4	NM_000539.3
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NC_000003.12:g.129532339A>G	ENST00000296271.4:c.619A>G	ENSP00000296271.3:p.Met207Val	missense_variant	RHO	ENSG00000163914	ENST00000296271.4	NM_000539.3
NC_000003.12:g.129532727C>A	ENST00000296271.4:c.891C>A	ENSP00000296271.3:p.Ser297Arg	missense_variant	RHO	ENSG00000163914	ENST00000296271.4	NM_000539.3
NC_000003.12:g.129533701C>T	ENST00000296271.4:c.1030C>T	ENSP00000296271.3:p.Gln344Ter	stop_gained	RHO	ENSG00000163914	ENST00000296271.4	NM_000539.3

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NC_000003.12:g.147413391C>A	ENST00000282928.5:c.1184C>A	ENSP00000282928.4:p.Pro395Gln	missense_variant	ZIC1	ENSG00000152977	ENST00000282928.5	NM_003412.4
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NC_000003.12:g.169107933C>A	ENST00000264674.7:c.2228G>T	ENSP00000264674.3:p.Arg743Ile	missense_variant	MECOM	ENSG00000085276	ENST00000264674.7	
NC_000003.12:g.169115722G>A	ENST00000264674.7:c.1781C>T	ENSP00000264674.3:p.Ser594Leu	missense_variant	MECOM	ENSG00000085276	ENST00000264674.7	
NC_000003.12:g.169116511G>A	ENST00000264674.7:c.992C>T	ENSP00000264674.3:p.Thr331Met	missense_variant	MECOM	ENSG00000085276	ENST00000264674.7	
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NC_000003.12:g.181712491C>G	ENST00000325404.3:c.131C>G	ENSP00000323588.1:p.Pro44Arg	missense_variant	SOX2	ENSG00000181449	ENST00000325404.3	NM_003106.4
NC_000003.12:g.181712565_181712566del	ENST00000325404.3:c.205_206del	ENSP00000323588.1:p.Ser69Glyfs	frameshift_variant	SOX2	ENSG00000181449	ENST00000325404.3	NM_003106.4
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NC_000004.12:g.1213028G>A	ENST00000290921.10:c.1024C>T	ENSP00000290921.6:p.Arg342Trp	missense_variant&splice_re	CTBP1	ENSG00000159692	ENST00000290921.10	
NC_000004.12:g.1803774C>A	ENST00000260795.8:c.*69C>A		3_prime_UTR_variant&NMI	FGFR3	ENSG00000068078	ENST00000260795.8	
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NC_000004.12:g.6301903G>A	ENST00000226760.5:c.2108G>A	ENSP00000226760.1:p.Arg703His	missense_variant	WFS1	ENSG00000109501	ENST00000226760.5	NM_006005.3
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NC_000004.12:g.82864362C>T	ENST00000264405.9:c.750G>A	ENSP00000264405.5:p.Lys250%3	splice_region_variant&syno	SEC31A	ENSG00000138674	ENST00000264405.9	
NC_000004.12:g.84702496A>G	ENST00000295888.9:c.8453T>C	ENSP00000295888.4:p.Phe2818S	missense_variant	WDFY3	ENSG00000163625	ENST00000295888.9	NM_014991.6
NC_000004.12:g.93829028dup	ENST00000306011.6:c.102dup	ENSP00000302216.4:p.Pro35Alafs	frameshift_variant	ATOH1	ENSG00000172238	ENST00000306011.6	NM_005172.2
NC_000004.12:g.95104467A>G	ENST00000264568.8:c.43A>G	ENSP00000264568.4:p.Lys15Glu	missense_variant	BMPR1B	ENSG00000138696	ENST00000264568.8	
NC_000004.12:g.99045192_99045193del	ENST00000296411.11:c.669_670del	ENSP00000296411.6:p.Tyr224Serf	frameshift_variant	METAP1	ENSG00000164024	ENST00000296411.11	NM_015143.3
NC_000004.12:g.108078308A>G	ENST00000265165.6:c.920T>C	ENSP00000265165.1:p.Met307Thr	missense_variant	LEF1	ENSG00000138795	ENST00000265165.6	NM_016269.5
NC_000004.12:g.112644733_112644734del	ENST00000344442.10:c.64_65del	ENSP00000344950.5:p.Glu22Serfs	frameshift_variant	LARP7	ENSG00000174720	ENST00000344442.10	NM_016648.4
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NC_000004.12:g.125452390C>T	ENST00000335110.5:c.6268C>T	ENSP00000335169.5:p.Arg2090Tr	missense_variant	FAT4	ENSG00000196159	ENST00000335110.5	
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NC_000004.12:g.152329692A>G	ENST00000281708.10:c.1216T>C	ENSP00000281708.3:p.Trp406Arg	missense_variant	FBXW7	ENSG00000109670	ENST00000281708.10	NM_001349798.2
NC_000004.12:g.186273168C>T	ENST00000403665.7:c.316C>T	ENSP00000384957.2:p.Gln106Ter	stop_gained	F11	ENSG00000088926	ENST00000403665.7	NM_000128.4
NC_000005.10:g.225989G>A	ENST00000264932.11:c.563G>A	ENSP00000264932.6:p.Arg188Gln	missense_variant	SDHA	ENSG00000073578	ENST00000264932.11	NM_004168.4
NC_000005.10:g.14374280A>G	ENST00000344204.9:c.3268A>G	ENSP00000339299.4:p.Arg1090Gl	missense_variant	TRIO	ENSG00000038382	ENST00000344204.9	NM_007118.4
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NC_000005.10:g.37064854C>T	ENST00000282516.13:c.8377C>T	ENSP00000282516.8:p.Arg2793Te	stop_gained	NIPBL	ENSG00000164190	ENST00000282516.13	NM_133433.4
NC_000005.10:g.37815889C>T	ENST00000326524.7:c.398G>A	ENSP00000317145.2:p.Gly133Asp	missense_variant	GDNF	ENSG00000168621	ENST00000326524.7	NM_000514.4
NC_000005.10:g.38502803T>C	ENST00000263409.8:c.1438-4A>G		splice_region_variant&splice	LIFR	ENSG00000113594	ENST00000263409.8	
NC_000005.10:g.38523416_38523419del	ENST00000263409.8:c.561+3_561+6del		splice_donor_variant&splice	LIFR	ENSG00000113594	ENST00000263409.8	
NC_000005.10:g.62381229del	ENST00000381103.7:c.1930del	ENSP00000370493.3:p.Ile644Ter	frameshift_variant	KIF2A	ENSG00000068796	ENST00000381103.7	
NC_000005.10:g.87363428C>T	ENST00000274376.11:c.1534C>T	ENSP00000274376.6:p.Arg512Ter	stop_gained	RASA1	ENSG00000145715	ENST00000274376.11	NM_002890.3
NC_000005.10:g.110743773C>T	ENST00000355943.8:c.370C>T	ENSP00000348211.3:p.Arg124Cys	missense_variant	SLC25A46	ENSG00000164209	ENST00000355943.8	NM_138773.4
NC_000005.10:g.110761328C>T	ENST00000355943.8:c.803C>T	ENSP00000348211.3:p.Thr268Met	missense_variant	SLC25A46	ENSG00000164209	ENST00000355943.8	NM_138773.4
NC_000005.10:g.127369927T>C	ENST00000274473.6:c.337T>C	ENSP00000274473.6:p.Cys113Arg	missense_variant	MEGF10	ENSG00000145794	ENST00000274473.6	
NC_000005.10:g.128309983C>G	ENST00000262464.9:c.5200G>C	ENSP00000262464.4:p.Asp1734H	missense_variant&splice_re	FBN2	ENSG00000138829	ENST00000262464.9	NM_001999.4
NC_000005.10:g.128312651C>T	ENST00000262464.9:c.4862G>A	ENSP00000262464.4:p.Cys1621Ty	missense_variant	FBN2	ENSG00000138829	ENST00000262464.9	NM_001999.4
NC_000005.10:g.132934208T>A	ENST00000265343.10:c.857A>T	ENSP00000265343.5:p.Glu286Val	missense_variant	AFF4	ENSG00000072364	ENST00000265343.10	NM_014423.4
NC_000005.10:g.135343259C>T	ENST00000304332.8:c.950+1G>A		splice_donor_variant	MACROH2A1	ENSG00000113648	ENST00000304332.8	
NC_000005.10:g.138427011_138427013dup	ENST00000314358.10:c.4448_4450dup	ENSP00000326563.5:p.Leu1483_I	inframe_insertion	KDM3B	ENSG00000120733	ENST00000314358.10	NM_016604.4
NC_000005.10:g.138782557A>G	ENST00000302763.12:c.105+528A>G		intron_variant	CTNNA1	ENSG00000044115	ENST00000302763.12	NM_001903.5
NC_000005.10:g.140114409C>G	ENST00000331327.5:c.228C>G	ENSP00000332706.3:p.Asp76Glu	missense_variant	PURA	ENSG00000185129	ENST00000331327.5	NM_005859.5

NC_000005.10:g.141526122G>A	ENST00000389054.8:c.3490C>T	ENSP00000373706.4:p.Arg1164Te	stop_gained	DIAPH1	ENSG00000131504	ENST00000389054.8	NM_005219.5
NC_000005.10:g.149883511C>T	ENST00000255266.10:c.2053G>A	ENSP00000255266.5:p.Val685Met	missense_variant	PDE6A	ENSG00000132915	ENST00000255266.10	NM_000440.3
NC_000005.10:g.149944370G>T	ENST00000255266.10:c.304C>A	ENSP00000255266.5:p.Arg102Ser	missense_variant	PDE6A	ENSG00000132915	ENST00000255266.10	NM_000440.3
NC_000005.10:g.150078240C>G	ENST00000286301.7:c.601G>C	ENSP00000286301.3:p.Gly201Arg	missense_variant	CSF1R	ENSG00000182578	ENST00000286301.7	
NC_000005.10:g.150357823C>T	ENST00000323668.11:c.77C>T	ENSP00000325223.6:p.Ala26Val	missense_variant	TCOF1	ENSG00000070814	ENST00000323668.11	
NC_000005.10:g.154888278G>A	ENST00000285873.8:c.4459C>T	ENSP00000285873.6:p.Gln1487Te	stop_gained	GEMIN5	ENSG00000082516	ENST00000285873.8	NM_015465.5
NC_000005.10:g.154899284C>T	ENST00000285873.8:c.3041G>A	ENSP00000285873.6:p.Arg1014Gl	missense_variant	GEMIN5	ENSG00000082516	ENST00000285873.8	NM_015465.5
NC_000005.10:g.154918006T>C	ENST00000285873.8:c.1600-2A>G		splice_acceptor_variant	GEMIN5	ENSG00000082516	ENST00000285873.8	NM_015465.5
NC_000005.10:g.173232939A>G	ENST00000329198.5:c.605T>C	ENSP00000327758.4:p.Leu202Prc	missense_variant	NKX2-5	ENSG00000183072	ENST00000329198.5	NM_004387.4
NC_000005.10:g.177269630C>T	ENST00000347982.9:c.4459C>T	ENSP00000343209.5:p.Arg1487Te	stop_gained	NSD1	ENSG00000165671	ENST00000347982.9	
NC_000005.10:g.177283815G>C	ENST00000347982.9:c.5165G>C	ENSP00000343209.5:p.Gly1722Al	missense_variant	NSD1	ENSG00000165671	ENST00000347982.9	
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NC_000005.10:g.177386233_177386253del	ENST00000324417.6:c.272_292del	ENSP00000321424.4:p.Val91_Ala	inframe_deletion	SLC34A1	ENSG00000131183	ENST00000324417.6	NM_003052.5
NC_000005.10:g.177397815G>A			downstream_gene_variant	F12	ENSG00000131187	ENST00000253496.4	NM_000505.4
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NC_000005.10:g.180620230C>A	ENST00000261937.11:c.2485G>T	ENSP00000261937.6:p.Glu829Ter	stop_gained	FLT4	ENSG00000037280	ENST00000261937.11	NM_182925.5
NC_000016.10:g.1566196_1566199del	ENST00000397417.6:c.*419_*422del		3_prime_UTR_variant&NMI	IFT140	ENSG00000187535	ENST00000397417.6	
NC_000006.12:g.3154297C>T	ENST00000333628.4:c.904G>A	ENSP00000369703.2:p.Ala302Thr	missense_variant	TUBB2A	ENSG00000137267	ENST00000333628.4	NM_001069.3
NC_000006.12:g.3154473G>A	ENST00000333628.4:c.728C>T	ENSP00000369703.2:p.Pro243Leu	missense_variant	TUBB2A	ENSG00000137267	ENST00000333628.4	NM_001069.3
NC_000006.12:g.3225348G>T	ENST00000259818.8:c.741C>A	ENSP00000259818.6:p.Asn247Lys	missense_variant	TUBB2B	ENSG00000137285	ENST00000259818.8	NM_178012.5
NC_000006.12:g.3227540G>C	ENST00000259818.8:c.4C>G	ENSP00000259818.6:p.Arg2Gly	missense_variant	TUBB2B	ENSG00000137285	ENST00000259818.8	NM_178012.5
NC_000006.12:g.21594710_21594711delins	ENST00000244745.4:c.176_177delinsAA	ENSP00000244745.1:p.Ile59Lys	missense_variant	SOX4	ENSG00000124766	ENST00000244745.4	NM_003107.3
NC_000006.12:g.21594739T>G	ENST00000244745.4:c.205T>G	ENSP00000244745.1:p.Trp69Gly	missense_variant	SOX4	ENSG00000124766	ENST00000244745.4	NM_003107.3
NC_000006.12:g.21595368dup	ENST00000244745.4:c.834dup	ENSP00000244745.1:p.Ala279Arg	frameshift_variant	SOX4	ENSG00000124766	ENST00000244745.4	NM_003107.3
NC_000006.12:g.32055923T>A	ENST00000375244.7:c.8395A>T	ENSP00000364393.3:p.Lys2799Te	stop_gained	TNXB	ENSG00000168477	ENST00000375244.7	
NC_000006.12:g.33166512C>T	ENST00000341947.7:c.4392+1G>A		splice_donor_variant	COL11A2	ENSG00000204248	ENST00000341947.7	NM_080680.3
NC_000006.12:g.33438775_33438794dup	ENST00000293748.9:c.1487_1506dup	ENSP00000293748.6:p.Tyr503Glu	frameshift_variant&splice_re	SYNGAP1	ENSG00000197283	ENST00000293748.9	
NC_000006.12:g.33692839C>T			downstream_gene_variant	UQCC2	ENSG00000137288	ENST00000374214.3	
NC_000006.12:g.42704570C>T	ENST00000230381.7:c.623G>A	ENSP00000230381.5:p.Gly208Asp	missense_variant	PRPH2	ENSG00000112619	ENST00000230381.7	NM_000322.5
NC_000006.12:g.42964370A>G	ENST00000244546.4:c.*444T>C		3_prime_UTR_variant	PEX6	ENSG00000124587	ENST00000244546.4	
NC_000006.12:g.43046047G>A	ENST00000265348.9:c.2705C>T	ENSP00000265348.4:p.Pro902Leu	missense_variant	CUL7	ENSG00000044090	ENST00000265348.9	NM_014780.5
NC_000006.12:g.45438040G>A	ENST00000359524.7:c.632G>A	ENSP00000352514.5:p.Arg211Gln	missense_variant	RUNX2	ENSG00000124813	ENST00000359524.7	
NC_000006.12:g.51659839T>A	ENST00000371117.8:c.10287A>T	ENSP00000360158.3:p.Pro3429%	synonymous_variant	PKHD1	ENSG00000170927	ENST00000371117.8	NM_138694.4
NC_000006.12:g.51755535T>C	ENST00000340994.4:c.8643-597A>G		intron_variant	PKHD1	ENSG00000170927	ENST00000340994.4	
NC_000006.12:g.51870590A>G	ENST00000340994.4:c.7400T>C	ENSP00000341097.4:p.Leu2467Pr	missense_variant	PKHD1	ENSG00000170927	ENST00000340994.4	
NC_000006.12:g.52002598C>G	ENST00000340994.4:c.5751+7711G>C		intron_variant	PKHD1	ENSG00000170927	ENST00000340994.4	
NC_000006.12:g.52070988G>A	ENST00000340994.4:c.667+18C>T		intron_variant	PKHD1	ENSG00000170927	ENST00000340994.4	
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NC_000006.12:g.80138817C>T	ENST00000320393.9:c.343+9588C>T		intron_variant	BCKDHB	ENSG00000083123	ENST00000320393.9	NM_183050.4
NC_000006.12:g.80191390A>G	ENST00000320393.9:c.743-9544A>G		intron_variant	BCKDHB	ENSG00000083123	ENST00000320393.9	NM_183050.4
NC_000006.12:g.85543737G>A	ENST00000314673.8:c.1132C>T	ENSP00000313121.3:p.Arg378Ter	stop_gained	SNX14	ENSG00000135317	ENST00000314673.8	NM_153816.6
NC_000006.12:g.87521502G>A	ENST00000369536.10:c.997C>T	ENSP00000358549.5:p.Arg333Ter	stop_gained	RARS2	ENSG00000146282	ENST00000369536.10	NM_020320.5
NC_000006.12:g.89950742T>C	ENST00000257749.9:c.1364A>G	ENSP00000257749.4:p.Asp455Gly	missense_variant	BACH2	ENSG00000112182	ENST00000257749.9	NM_021813.4
NC_000006.12:g.104744197_104744199del	ENST00000262903.9:c.2475_2477del	ENSP00000262903.4:p.Glu827del	inframe_deletion	HACE1	ENSG00000085382	ENST00000262903.9	NM_020771.4
NC_000006.12:g.112068342_112068346del	ENST00000230529.9:c.727_731del	ENSP00000230529.5:p.Glu243Lys	frameshift_variant	CCN6	ENSG00000112761	ENST00000230529.9	
NC_000006.12:g.121446966C>T	ENST00000282561.4:c.119C>T	ENSP00000282561.3:p.Ala40Val	missense_variant	GJA1	ENSG00000152661	ENST00000282561.4	NM_000165.5
NC_000006.12:g.121447022C>T	ENST00000282561.4:c.175C>T	ENSP00000282561.3:p.Pro59Ser	missense_variant	GJA1	ENSG00000152661	ENST00000282561.4	NM_000165.5
NC_000006.12:g.122717874G>A	ENST00000258014.3:c.101G>A	ENSP00000258014.3:p.Arg34Gln	missense_variant	PKIB	ENSG00000135549	ENST00000258014.3	
NC_000006.12:g.129252253T>C	ENST00000421865.3:c.2054T>C	ENSP00000400365.2:p.Leu685Prc	missense_variant	LAMA2	ENSG00000196569	ENST00000421865.3	NM_000426.4
NC_000006.12:g.129402492G>A	ENST00000421865.3:c.5726+5G>A		splice_donor_5th_base_var	LAMA2	ENSG00000196569	ENST00000421865.3	NM_000426.4
NC_000006.12:g.136869909C>T	ENST00000318471.5:c.653C>T	ENSP00000315680.3:p.Ala218Val	missense_variant	PEX7	ENSG00000112357	ENST00000318471.5	NM_000288.4
NC_000006.12:g.136898192A>G	ENST00000318471.5:c.854A>G	ENSP00000315680.3:p.His285Arg	missense_variant	PEX7	ENSG00000112357	ENST00000318471.5	NM_000288.4
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NC_000006.12:g.151430154T>C	ENST00000336451.8:c.*112A>G		3_prime_UTR_variant&NMI	RMND1	ENSG00000155906	ENST00000336451.8	
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NC_000006.12:g.152350316_152350319del	ENST00000367255.10:c.11750_11753del	ENSP00000356224.5:p.Leu3917Al	frameshift_variant	SYNE1	ENSG00000131018	ENST00000367255.10	NM_182961.4
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NC_000007.14:g.5529399C>G	ENST00000414620.1:c.125G>C	ENSP00000401032.1:p.Gly42Ala	missense_variant&splice_re	ACTB	ENSG00000075624	ENST00000414620.1	
NC_000007.14:g.6009436T>C	ENST00000223029.8:c.73T>C	ENSP00000223029.3:p.Tyr25His	missense_variant	AIMP2	ENSG00000106305	ENST00000223029.8	NM_006303.4

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NC_000007.14:g.16376237G>A	ENST00000399310.3:c.534+29824C>T		intron_variant	CRPPA	ENSG00000214960	ENST00000399310.3	
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NC_000007.14:g.39997498C>T	ENST00000181839.10:c.1876C>T	ENSP00000181839.4:p.Arg626Ter	stop_gained	CDK13	ENSG00000065883	ENST00000181839.10	NM_003718.5
NC_000007.14:g.40001924C>A	ENST00000181839.10:c.2246C>A	ENSP00000181839.4:p.Ala749Glu	missense_variant	CDK13	ENSG00000065883	ENST00000181839.10	NM_003718.5
NC_000007.14:g.40046007A>G	ENST00000181839.10:c.2525A>G	ENSP00000181839.4:p.Asn842Ser	missense_variant	CDK13	ENSG00000065883	ENST00000181839.10	NM_003718.5
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NC_000007.14:g.41966180C>A	ENST00000395925.8:c.2893G>T	ENSP00000379258.3:p.Glu965Ter	stop_gained	GLI3	ENSG00000106571	ENST00000395925.8	NM_000168.6
NC_000007.14:g.41967653G>A	ENST00000395925.8:c.2374C>T	ENSP00000379258.3:p.Arg792Ter	stop_gained	GLI3	ENSG00000106571	ENST00000395925.8	NM_000168.6
NC_000007.14:g.47831215_47831216del	ENST00000289672.7:c.6473+2_6473+3del		splice_donor_variant	PKD1L1	ENSG00000158683	ENST00000289672.7	NM_138295.5
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NC_000007.14:g.66994210A>G	ENST00000246868.7:c.258+2T>C		splice_donor_variant	SBDS	ENSG00000126524	ENST00000246868.7	NM_016038.4
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NC_000007.14:g.66994286T>A	ENST00000246868.7:c.184A>T	ENSP00000246868.2:p.Lys62Ter	stop_gained	SBDS	ENSG00000126524	ENST00000246868.7	NM_016038.4
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NC_000007.14:g.70766256C>G	ENST00000342771.10:c.1611C>G	ENSP00000344087.4:p.His537Gln	missense_variant	AUTS2	ENSG00000158321	ENST00000342771.10	NM_015570.4
NC_000007.14:g.74046204del	ENST00000252034.12:c.558del	ENSP00000252034.7:p.Phe186Leu	frameshift_variant	ELN	ENSG00000049540	ENST00000252034.12	NM_000501.4
NC_000007.14:g.74053251C>T	ENST00000252034.12:c.1038C>T	ENSP00000252034.7:p.Val346%3D	synonymous_variant	ELN	ENSG00000049540	ENST00000252034.12	NM_000501.4
NC_000007.14:g.74066774del	ENST00000252034.12:c.2129del	ENSP00000252034.7:p.Pro710Gln	frameshift_variant&splice_re	ELN	ENSG00000049540	ENST00000252034.12	NM_000501.4
NC_000007.14:g.76057482C>G	ENST00000315758.10:c.308C>G	ENSP00000327070.5:p.Pro103Arg	missense_variant	MDH2	ENSG00000146701	ENST00000315758.10	NM_005918.4
NC_000007.14:g.93103784C>T	ENST00000379958.3:c.2314G>A	ENSP00000369292.2:p.Glu772Lys	missense_variant	SAMD9	ENSG00000205413	ENST00000379958.3	NM_017654.4
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NC_000007.14:g.107672245G>T			downstream_gene_variant	SLC26A4	ENSG00000091137	ENST00000440056.1	
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NC_000007.14:g.116898838A>G	ENST00000361183.8:c.219+3A>G		splice_donor_region_varian	CAPZA2	ENSG00000198898	ENST00000361183.8	NM_006136.3
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NC_000007.14:g.140119149A>C	ENST00000397560.7:c.1210T>G	ENSP00000380692.2:p.Trp404Gly	missense_variant	KDM7A	ENSG00000006459	ENST00000397560.7	NM_030647.2
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NC_000008.11:g.42870016G>A	ENST00000240159.8:c.310C>T	ENSP00000240159.4:p.His104Tyr	missense_variant&NMD_tra	RNF170	ENSG00000120925	ENST00000240159.8	
NC_000008.11:g.43197910G>C	ENST00000379644.9:c.1684G>C	ENSP00000368965.4:p.Asp562His	missense_variant	HGSNAT	ENSG00000165102	ENST00000379644.9	NM_152419.3
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NC_000008.11:g.54621572C>A	ENST00000220676.2:c.606C>A	ENSP00000220676.1:p.Asp202Glu	missense_variant	RP1	ENSG00000104237	ENST00000220676.2	NM_006269.2
NC_000008.11:g.54624669G>A	ENST00000220676.2:c.788-1G>A		splice_acceptor_variant	RP1	ENSG00000104237	ENST00000220676.2	NM_006269.2
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NC_000008.11:g.54627701_54627702del	ENST00000220676.2:c.3819_3820del	ENSP00000220676.1:p.Cys1274Te	frameshift_variant	RP1	ENSG00000104237	ENST00000220676.2	NM_006269.2
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NC_000008.11:g.67179927G>A	ENST00000262210.11:c.3286+1G>A		splice_donor_variant	CSPP1	ENSG00000104218	ENST00000262210.11	
NC_000008.11:g.71211155C>T	ENST00000303824.11:c.1680+1G>A		splice_donor_variant	EYA1	ENSG00000104313	ENST00000303824.11	
NC_000008.11:g.71215630A>G	ENST00000303824.11:c.1441T>C	ENSP00000303221.7:p.Ser481Pro	missense_variant	EYA1	ENSG00000104313	ENST00000303824.11	
NC_000008.11:g.71269734A>G	ENST00000303824.11:c.1032+6T>C		splice_donor_region_varian	EYA1	ENSG00000104313	ENST00000303824.11	

NC_000008.11:g.71271778dup	ENST00000303824.11:c.932dup	ENSP00000303221.7:p.Asp312Arg	frameshift_variant	EYA1	ENSG00000104313	ENST00000303824.11	
NC_000008.11:g.79765563C>T	ENST00000337919.9:c.552G>A	ENSP00000338272.5:p.Trp184Ter	stop_gained	HEY1	ENSG00000164683	ENST00000337919.9	
NC_000008.11:g.86643781del	ENST00000320005.6:c.1148del	ENSP00000316605.5:p.Thr383Ilefs	frameshift_variant	CNGB3	ENSG00000170289	ENST00000320005.6	NM_019098.5
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NC_000008.11:g.93815442del	ENST00000323130.8:c.2902del	ENSP00000314488.4:p.Gln968Lys	frameshift_variant&NMD_tra	TMEM67	ENSG00000164953	ENST00000323130.8	
NC_000008.11:g.103318623_103318625del	ENST00000358755.5:c.211_213del	ENSP00000351605.4:p.Pro71del	inframe_deletion	FZD6	ENSG00000164930	ENST00000358755.5	NM_003506.4
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NC_000008.11:g.142874997C>T	ENST00000292427.10:c.1358G>A	ENSP00000292427.5:p.Arg453Gln	missense_variant	CYP11B1	ENSG00000160882	ENST00000292427.10	NM_000497.4
NC_000008.11:g.142876840_142876841del	ENST00000292427.10:c.642_643del	ENSP00000292427.5:p.His214Gln	frameshift_variant	CYP11B1	ENSG00000160882	ENST00000292427.10	NM_000497.4
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NC_000009.12:g.95477549G>A	ENST00000331920.11:c.1501C>T	ENSP00000332353.6:p.Gln501Ter	stop_gained&splice_region_	PTCH1	ENSG00000185920	ENST00000331920.11	NM_000264.5
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NC_000009.12:g.132298269_132298270del	ENST00000224140.6:c.5591_5592del	ENSP00000224140.5:p.Gln1864Ar	frameshift_variant	SETX	ENSG00000107290	ENST00000224140.6	NM_015046.7
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NC_000009.12:g.136417149T>C	ENST00000371717.8:c.832T>C	ENSP00000360782.3:p.Trp278Arg	missense_variant	PMPCA	ENSG00000165688	ENST00000371717.8	NM_015160.3
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NC_000009.12:g.136432980G>A	ENST00000371712.4:c.1255C>T	ENSP00000360777.3:p.Leu419Phe	missense_variant	INPP5E	ENSG00000148384	ENST00000371712.4	NM_019892.6
NC_000009.12:g.136434825A>G	ENST00000371712.4:c.851T>C	ENSP00000360777.3:p.Leu284Pro	missense_variant	INPP5E	ENSG00000148384	ENST00000371712.4	NM_019892.6
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NC_000009.12:g.136514510C>G			upstream_gene_variant	NOTCH1	ENSG00000148400	ENST00000645828.1	
NC_000009.12:g.136515369A>T			downstream_gene_variant	MIR4673	ENSG00000263403	ENST00000584777.1	
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NC_000009.12:g.136537613T>C	ENST00000651671.1:c.140+6411A>G		intron_variant	NOTCH1	ENSG00000148400	ENST00000651671.1	NM_017617.5
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NC_000009.12:g.137243390G>A	ENST00000340384.5:c.1172G>A	ENSP00000341289.4:p.Arg391His	missense_variant	TUBB4B	ENSG00000188229	ENST00000340384.5	NM_006088.6
NC_000009.12:g.137776723G>T	ENST00000371394.6:c.*1632G>T		3_prime_UTR_variant&NMI	EHMT1	ENSG00000181090	ENST00000371394.6	
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NC_000010.11:g.69015836T>G	ENST00000361983.7:c.1286T>G	ENSP00000354848.4:p.Leu429Arg	missense_variant	KIFBP	ENSG00000198954	ENST00000361983.7	NM_015634.4
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NC_000010.11:g.78009682G>C	ENST00000372371.8:c.1771-7C>G		splice_region_variant&splice	POLR3A	ENSG00000148606	ENST00000372371.8	NM_007055.4
NC_000010.11:g.79292262T>G	ENST00000334512.10:c.863T>G	ENSP00000334474.5:p.Val288Gly	missense_variant	ZMIZ1	ENSG00000108175	ENST00000334512.10	NM_020338.4
NC_000010.11:g.87504973G>T	ENST00000371994.8:c.58G>T	ENSP00000361062.4:p.Ala20Ser	missense_variant	MINPP1	ENSG00000107789	ENST00000371994.8	
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NC_000010.11:g.110580974T>C	ENST00000361804.5:c.500T>C	ENSP00000354720.5:p.Val167Ala	missense_variant	SMC3	ENSG00000108055	ENST00000361804.5	NM_005445.4
NC_000010.11:g.114132185C>G	ENST00000369287.8:c.1656-1G>C		splice_acceptor_variant	CCDC186	ENSG00000165813	ENST00000369287.8	NM_018017.4
NC_000010.11:g.119146121_119146811del	ENST00000355697.7:c.819-458_936+115del		splice_acceptor_variant&spl	SFXN4	ENSG00000183605	ENST00000355697.7	NM_213649.2
NC_000010.11:g.119157870C>T	ENST00000355697.7:c.471+1G>A		splice_donor_variant	SFXN4	ENSG00000183605	ENST00000355697.7	NM_213649.2
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NC_000010.11:g.129873521A>G	ENST00000355311.10:c.712T>C	ENSP00000347463.4:p.Ser238Pro	missense_variant	EBF3	ENSG00000108001	ENST00000355311.10	
NC_000010.11:g.129873547G>A	ENST00000355311.10:c.686C>T	ENSP00000347463.4:p.Ser229Leu	missense_variant	EBF3	ENSG00000108001	ENST00000355311.10	
NC_000010.11:g.129877779G>A	ENST00000355311.10:c.625C>T	ENSP00000347463.4:p.Arg209Trp	missense_variant	EBF3	ENSG00000108001	ENST00000355311.10	
NC_000010.11:g.133369929A>T	ENST00000368547.4:c.389T>A	ENSP00000357535.3:p.Val130Asp	missense_variant	ECHS1	ENSG00000127884	ENST00000368547.4	NM_004092.4
NC_000010.11:g.133370578C>T	ENST00000368547.4:c.268G>A	ENSP00000357535.3:p.Gly90Arg	missense_variant	ECHS1	ENSG00000127884	ENST00000368547.4	NM_004092.4
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NC_000011.10:g.755962C>G	ENST00000319006.8:c.181C>G	ENSP00000321259.3:p.Leu61Val	missense_variant	TALDO1	ENSG00000177156	ENST00000319006.8	NM_006755.2
NC_000011.10:g.795413_795421dup	ENST00000320230.9:c.-163-247_-163-239dup		intron_variant	SLC25A22	ENSG00000177542	ENST00000320230.9	
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NC_000011.10:g.2135424C>T	ENST00000381389.5:c.100G>A	ENSP00000370796.1:p.Gly34Ser	missense_variant	IGF2	ENSG00000167244	ENST00000381389.5	
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NC_000011.10:g.13728665G>A	ENST00000354817.8:c.1439G>A	ENSP00000346874.3:p.Arg480His	missense_variant	FAR1	ENSG00000197601	ENST00000354817.8	NM_032228.6
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NC_000011.10:g.61781776C>T	ENST00000265460.9:c.2863C>T	ENSP00000265460.5:p.Gln955Ter	stop_gained	MYRF	ENSG00000124920	ENST00000265460.9	
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NC_000011.10:g.65346060dup	ENST00000252268.8:c.946+2dup		splice_donor_variant	DPF2	ENSG00000133884	ENST00000252268.8	
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NC_000011.10:g.66705399A>G	ENST00000309996.7:c.1877T>C	ENSP00000311489.2:p.Leu626Prc	missense_variant	SPTBN2	ENSG00000173898	ENST00000309996.7	
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NC_000011.10:g.108299461C>G	ENST00000278616.10:c.5006-253C>G		intron_variant	ATM	ENSG00000149311	ENST00000278616.10	
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NC_000011.10:g.117390886C>T	ENST00000278935.8:c.2044C>T	ENSP00000278935.3:p.Gln682Ter	stop_gained	CEP164	ENSG00000110274	ENST00000278935.8	NM_014956.5
NC_000011.10:g.118480180A>T	ENST00000389506.10:c.3576A>T	ENSP00000374157.5:p.Arg1192Se	missense_variant	KMT2A	ENSG00000118058	ENST00000389506.10	
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NC_000011.10:g.124868850C>G	ENST00000397801.6:c.209C>G	ENSP00000380903.1:p.Pro70Arg	missense_variant	ROBO3	ENSG00000154134	ENST00000397801.6	NM_022370.4
NC_000011.10:g.128839370G>A	ENST00000324036.7:c.874C>T	ENSP00000316233.3:p.Arg292Trp	missense_variant	KCNJ1	ENSG00000151704	ENST00000324036.7	
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NC_000014.9:g.101964717_101964719del	ENST00000360184.10:c.26_28del	ENSP00000348965.4:p.Gly9del	inframe_deletion	DYNC1H1	ENSG00000197102	ENST00000360184.10	NM_001376.5
NC_000014.9:g.101979951C>T	ENST00000360184.10:c.751C>T	ENSP00000348965.4:p.Arg251Cys	missense_variant	DYNC1H1	ENSG00000197102	ENST00000360184.10	NM_001376.5
NC_000014.9:g.102029936_102029938del	ENST00000360184.10:c.9760_9762del	ENSP00000348965.4:p.Lys3254de	inframe_deletion&splice_reg	DYNC1H1	ENSG00000197102	ENST00000360184.10	NM_001376.5
NC_000015.10:g.23645753dup	ENST00000650528.1:c.1996dup	ENSP00000497810.1:p.Gln666Pro	frameshift_variant	MAGEL2	ENSG00000254585	ENST00000650528.1	NM_019066.5
NC_000015.10:g.23647647_23647724dup	ENST00000650528.1:c.19_96dup	ENSP00000497810.1:p.Asn7_Ala3	inframe_insertion	MAGEL2	ENSG00000254585	ENST00000650528.1	NM_019066.5
NC_000015.10:g.28233184A>G	ENST00000261609.13:c.4637T>C	ENSP00000261609.8:p.Ile1546Thr	missense_variant	HERC2	ENSG00000128731	ENST00000261609.13	NM_004667.6
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NC_000015.10:g.31063252del	ENST00000256552.11:c.836del	ENSP00000256552.7:p.Gly279Alaf	frameshift_variant	TRPM1	ENSG00000134160	ENST00000256552.11	NM_001252024.2
NC_000015.10:g.31068091T>C	ENST00000256552.11:c.281A>G	ENSP00000256552.7:p.Tyr94Cys	missense_variant&splice_re	TRPM1	ENSG00000134160	ENST00000256552.11	NM_001252024.2
NC_000015.10:g.34792177T>A	ENST00000290378.6:c.721A>T	ENSP00000290378.4:p.Ser241Cys	missense_variant	ACTC1	ENSG00000159251	ENST00000290378.6	NM_005159.5
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NC_000015.10:g.34793341T>G	ENST00000290378.6:c.358A>C	ENSP00000290378.4:p.Lys120Gln	missense_variant	ACTC1	ENSG00000159251	ENST00000290378.6	NM_005159.5
NC_000015.10:g.34793439A>G	ENST00000290378.6:c.260T>C	ENSP00000290378.4:p.Ile87Thr	missense_variant	ACTC1	ENSG00000159251	ENST00000290378.6	NM_005159.5
NC_000015.10:g.36896639G>T	ENST00000314177.12:c.*39C>A		3_prime_UTR_variant&NMD	MEIS2	ENSG00000134138	ENST00000314177.12	
NC_000015.10:g.48429426G>C	ENST00000316623.10:c.6872-955C>G		intron_variant	FBN1	ENSG00000166147	ENST00000316623.10	NM_000138.5
NC_000015.10:g.48613060T>C	ENST00000316623.10:c.197A>G	ENSP00000325527.5:p.Tyr66Cys	missense_variant	FBN1	ENSG00000166147	ENST00000316623.10	NM_000138.5
NC_000015.10:g.51487998C>G	ENST00000251076.9:c.5173G>C	ENSP00000251076.5:p.Ala1725Pr	missense_variant	DMXL2	ENSG00000104093	ENST00000251076.9	
NC_000015.10:g.56444534dup	ENST00000260453.4:c.605dup	ENSP00000260453.3:p.Gln203Ala	frameshift_variant	MNS1	ENSG00000138587	ENST00000260453.4	NM_018365.4
NC_000015.10:g.56464182_56464183delins	ENST00000260453.4:c.68_69delinsAG	ENSP00000260453.3:p.Cys23Ter	stop_gained	MNS1	ENSG00000138587	ENST00000260453.4	NM_018365.4
NC_000015.10:g.57232402del	ENST00000267811.9:c.797del	ENSP00000267811.5:p.Tyr266Leu	frameshift_variant	TCF12	ENSG00000140262	ENST00000267811.9	
NC_000015.10:g.68211685G>A	ENST00000249806.11:c.476C>T	ENSP00000249806.5:p.Pro159Leu	missense_variant	CLN6	ENSG00000128973	ENST00000249806.11	NM_017882.3
NC_000015.10:g.72349153_72349155del	ENST00000268097.10:c.915_917del	ENSP00000268097.6:p.Phe305del	inframe_deletion	HEXA	ENSG00000213614	ENST00000268097.10	NM_000520.6
NC_000015.10:g.72731573C>T	ENST00000268057.9:c.883C>T	ENSP00000268057.4:p.Arg295Ter	stop_gained	BBS4	ENSG00000140463	ENST00000268057.9	NM_033028.5
NC_000015.10:g.74180923G>A			downstream_gene_variant	ISLR	ENSG00000129009	ENST00000249842.8	NM_005545.4
NC_000015.10:g.74190853G>A	ENST00000323940.9:c.914C>T	ENSP00000326085.5:p.Thr305Met	missense_variant	STRA6	ENSG00000137868	ENST00000323940.9	
NC_000015.10:g.75392256C>T	ENST00000360439.8:c.2837G>A	ENSP00000353622.4:p.Arg946His	missense_variant	SIN3A	ENSG00000169375	ENST00000360439.8	
NC_000015.10:g.76311367_76311377dup	ENST00000267950.12:c.15_25dup	ENSP00000267950.8:p.Gln9Argfs	frameshift_variant&NMD_tra	ETFA	ENSG00000140374	ENST00000267950.12	
NC_000015.10:g.88836207A>G	ENST00000352105.11:c.1A>G	ENSP00000341615.7:p.Met1?	start_lost	ACAN	ENSG00000157766	ENST00000352105.11	
NC_000015.10:g.89628466G>A			downstream_gene_variant	TICRR	ENSG00000140534	ENST00000268138.12	NM_152259.4
NC_000015.10:g.89631709_89631710del			downstream_gene_variant	TICRR	ENSG00000140534	ENST00000268138.12	NM_152259.4
NC_000015.10:g.100341183G>C	ENST00000268070.9:c.306C>G	ENSP00000268070.4:p.Phe102Leu	missense_variant	ADAMTS17	ENSG00000140470	ENST00000268070.9	NM_139057.4
NC_000015.10:g.101021900C>T	ENST00000388948.8:c.1795C>T	ENSP00000373600.3:p.Gln599Ter	stop_gained	LRRK1	ENSG00000154237	ENST00000388948.8	NM_024652.6
NC_000016.10:g.2102428C>G	ENST00000262304.9:c.9154G>C	ENSP00000262304.4:p.Gly3052Ar	missense_variant	PKD1	ENSG00000008710	ENST00000262304.9	NM_001009944.3
NC_000016.10:g.173001T>C	ENST00000251595.11:c.89T>C	ENSP00000251595.6:p.Leu30Pro	missense_variant	HBA2	ENSG00000188536	ENST00000251595.11	NM_000517.6
NC_000016.10:g.680671A>G	ENST00000219548.9:c.146A>G	ENSP00000219548.4:p.Tyr49Cys	missense_variant	STUB1	ENSG00000103266	ENST00000219548.9	NM_005861.4
NC_000016.10:g.1449887A>T	ENST00000262318.12:c.1546-560T>A		intron_variant	CLCN7	ENSG00000103249	ENST00000262318.12	
NC_000016.10:g.1583313C>T	ENST00000397417.6:c.605+1G>A		splice_donor_variant&NMD_	IFT140	ENSG00000187535	ENST00000397417.6	
NC_000016.10:g.2103784_2103790dup	ENST00000262304.9:c.8271_8277dup	ENSP00000262304.4:p.Met2760C	frameshift_variant	PKD1	ENSG00000008710	ENST00000262304.9	NM_001009944.3
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NC_000016.10:g.2109988G>C	ENST00000262304.9:c.5179C>G	ENSP00000262304.4:p.Pro1727Al	missense_variant	PKD1	ENSG00000008710	ENST00000262304.9	NM_001009944.3
NC_000016.10:g.2176153C>G	ENST00000326181.11:c.1851C>G	ENSP00000318944.6:p.Phe617Leu	missense_variant	TRAF7	ENSG00000131653	ENST00000326181.11	NM_032271.3
NC_000016.10:g.2464233del	ENST00000361837.9:c.1155+4del		splice_donor_region_varian	TEDC2	ENSG00000162062	ENST00000361837.9	NM_025108.3
NC_000016.10:g.2496476G>A	ENST00000562105.2:c.328G>A	ENSP00000457896.2:p.Gly110Ser	missense_variant	TBC1D24	ENSG00000162065	ENST00000562105.2	
NC_000016.10:g.2496587G>C	ENST00000562105.2:c.439G>C	ENSP00000457896.2:p.Asp147His	missense_variant	TBC1D24	ENSG00000162065	ENST00000562105.2	
NC_000016.10:g.2496705del	ENST00000562105.2:c.557del	ENSP00000457896.2:p.Leu186Arg	frameshift_variant	TBC1D24	ENSG00000162065	ENST00000562105.2	
NC_000016.10:g.2496788C>T	ENST00000562105.2:c.640C>T	ENSP00000457896.2:p.Arg214Cys	missense_variant	TBC1D24	ENSG00000162065	ENST00000562105.2	
NC_000016.10:g.2496957G>A	ENST00000562105.2:c.809G>A	ENSP00000457896.2:p.Arg270His	missense_variant	TBC1D24	ENSG00000162065	ENST00000562105.2	
NC_000016.10:g.2497433G>A	ENST00000562105.2:c.966-277G>A		intron_variant	TBC1D24	ENSG00000162065	ENST00000562105.2	
NC_000016.10:g.3767734T>G	ENST00000262367.10:c.3236A>C	ENSP00000262367.5:p.Gln1079Pr	missense_variant	CREBBP	ENSG00000005339	ENST00000262367.10	NM_004380.3
NC_000016.10:g.13926729_13926730del	ENST00000311895.8:c.557_558del	ENSP00000310520.7:p.Phe186Cys	frameshift_variant	ERCC4	ENSG00000175595	ENST00000311895.8	NM_005236.3
NC_000016.10:g.13947991C>T	ENST00000311895.8:c.2395C>T	ENSP00000310520.7:p.Arg799Trp	missense_variant	ERCC4	ENSG00000175595	ENST00000311895.8	NM_005236.3
NC_000016.10:g.18798759G>A	ENST00000304414.12:c.112C>T	ENSP00000306788.7:p.Arg38Ter	stop_gained	ARL6IP1	ENSG00000170540	ENST00000304414.12	NM_015161.3
NC_000016.10:g.20348552T>A	ENST00000396134.6:c.848A>T	ENSP00000379438.2:p.His283Leu	missense_variant	UMOD	ENSG00000169344	ENST00000396134.6	
NC_000016.10:g.23628674C>T	ENST00000261584.9:c.2586+530G>A		intron_variant	PALB2	ENSG00000083093	ENST00000261584.9	NM_024675.4
NC_000016.10:g.27210506_27210518del	ENST00000286096.9:c.383_395del	ENSP00000286096.5:p.Leu128Prc	frameshift_variant	KDM8	ENSG00000155666	ENST00000286096.9	NM_024773.3
NC_000016.10:g.28482500C>T	ENST00000333496.14:c.811G>A	ENSP00000329171.9:p.Glu271Lys	missense_variant	CLN3	ENSG00000188603	ENST00000333496.14	
NC_000016.10:g.28844035C>T	ENST00000313511.8:c.989G>A	ENSP00000322439.3:p.Arg330Gln	missense_variant	TUFM	ENSG00000178952	ENST00000313511.8	NM_003321.5
NC_000016.10:g.28844287T>C	ENST00000313511.8:c.865A>G	ENSP00000322439.3:p.Lys289Glu	missense_variant	TUFM	ENSG00000178952	ENST00000313511.8	NM_003321.5
NC_000016.10:g.29798639C>G	ENST00000160827.9:c.441C>G	ENSP00000160827.5:p.Ile147Met	missense_variant	KIF22	ENSG00000079616	ENST00000160827.9	NM_007317.3
NC_000016.10:g.30704236T>A	ENST00000262518.9:c.227T>A	ENSP00000262518.4:p.Leu76Ter	stop_gained	SRCAP	ENSG00000080603	ENST00000262518.9	NM_006662.3
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NC_000016.10:g.30980766C>T	ENST00000262519.14:c.4609C>T	ENSP00000262519.8:p.Arg1537Tr	missense_variant	SETD1A	ENSG00000099381	ENST00000262519.14	NM_014712.3

NC_000016.10:g.55491894A>C	ENST00000219070.9:c.1274A>C	ENSP00000219070.4:p.Tyr425Ser	missense_variant	MMP2	ENSG00000087245	ENST00000219070.9	NM_004530.6
NC_000016.10:g.56336762C>T	ENST00000262493.12:c.625C>T	ENSP00000262493.6:p.Arg209Cys	missense_variant	GNAO1	ENSG00000087258	ENST00000262493.12	NM_020988.3
NC_000016.10:g.56506181T>C	ENST00000245157.11:c.656A>G	ENSP00000245157.5:p.Tyr219Cys	missense_variant	BBS2	ENSG00000125124	ENST00000245157.11	NM_031885.5
NC_000016.10:g.56519795C>G	ENST00000245157.11:c.68G>C	ENSP00000245157.5:p.Arg23Pro	missense_variant	BBS2	ENSG00000125124	ENST00000245157.11	NM_031885.5
NC_000016.10:g.57887971T>A	ENST00000251102.13:c.3346A>T	ENSP00000251102.8:p.Thr1116Se	missense_variant	CNGB1	ENSG00000070729	ENST00000251102.13	NM_001297.5
NC_000016.10:g.57904865A>G	ENST00000251102.13:c.2503T>C	ENSP00000251102.8:p.Cys835Arg	missense_variant	CNGB1	ENSG00000070729	ENST00000251102.13	NM_001297.5
NC_000016.10:g.67066713G>A	ENST00000290858.11:c.314G>A	ENSP00000290858.6:p.Gly105Glu	missense_variant	CBFB	ENSG00000067955	ENST00000290858.11	
NC_000016.10:g.67621577G>A	ENST00000264010.10:c.1343G>A	ENSP00000264010.4:p.Arg448Gln	missense_variant	CTCF	ENSG00000102974	ENST00000264010.10	NM_006565.4
NC_000016.10:g.68808700C>T	ENST00000261769.10:c.539C>T	ENSP00000261769.4:p.Ser180Phe	missense_variant	CDH1	ENSG00000039068	ENST00000261769.10	NM_004360.5
NC_000016.10:g.69320768A>T	ENST00000254950.13:c.850A>T	ENSP00000254950.11:p.Arg284Tr	missense_variant&splice_re	VPS4A	ENSG00000132612	ENST00000254950.13	NM_013245.3
NC_000016.10:g.70698720T>C	ENST00000261776.10:c.1753A>G	ENSP00000261776.5:p.Met585Val	missense_variant	VAC14	ENSG00000103043	ENST00000261776.10	NM_018052.5
NC_000016.10:g.70698834T>C	ENST00000261776.10:c.1662-23A>G		intron_variant	VAC14	ENSG00000103043	ENST00000261776.10	NM_018052.5
NC_000016.10:g.75545891G>C	ENST00000258173.11:c.373C>G	ENSP00000258173.5:p.Pro125Ala	missense_variant	TMEM231	ENSG00000205084	ENST00000258173.11	NM_001077418.3
NC_000016.10:g.75556139G>T	ENST00000258173.11:c.71C>A	ENSP00000258173.5:p.Ala24Glu	missense_variant	TMEM231	ENSG00000205084	ENST00000258173.11	NM_001077418.3
NC_000016.10:g.79599026T>C	ENST00000326043.5:c.877A>G	ENSP00000327048.4:p.Arg293Gly	missense_variant	MAF	ENSG00000178573	ENST00000326043.5	NM_005360.5
NC_000016.10:g.88719588G>A	ENST00000301015.14:c.6457C>T	ENSP00000301015.9:p.Arg2153Te	stop_gained	PIEZO1	ENSG00000103335	ENST00000301015.14	NM_001142864.4
NC_000016.10:g.88742096C>T	ENST00000301015.14:c.284-1G>A		splice_acceptor_variant	PIEZO1	ENSG00000103335	ENST00000301015.14	NM_001142864.4
NC_000016.10:g.88814540T>C	ENST00000268695.10:c.1483-15A>G		splice_polypyrimidine_tract_	GALNS	ENSG00000141012	ENST00000268695.10	NM_000512.5
NC_000016.10:g.88822674C>A	ENST00000268695.10:c.1279G>T	ENSP00000268695.5:p.Val427Phe	missense_variant	GALNS	ENSG00000141012	ENST00000268695.10	NM_000512.5
NC_000016.10:g.89101154C>T	ENST00000317447.9:c.473C>T	ENSP00000320646.4:p.Pro158Leu	missense_variant	ACSF3	ENSG00000176715	ENST00000317447.9	
NC_000016.10:g.89102665C>T	ENST00000317447.9:c.728C>T	ENSP00000320646.4:p.Pro243Leu	missense_variant	ACSF3	ENSG00000176715	ENST00000317447.9	
NC_000016.10:g.89270888G>A	ENST00000301030.10:c.7735C>T	ENSP00000301030.4:p.Arg2579Cy	missense_variant	ANKRD11	ENSG00000167522	ENST00000301030.10	NM_013275.6
NC_000016.10:g.89282415_89282416dup	ENST00000301030.10:c.4126_4127dup	ENSP00000301030.4:p.Gly1377Ar	frameshift_variant	ANKRD11	ENSG00000167522	ENST00000301030.10	NM_013275.6
NC_000016.10:g.89282772_89282773del	ENST00000301030.10:c.3770_3771del	ENSP00000301030.4:p.Lys1257Ar	frameshift_variant	ANKRD11	ENSG00000167522	ENST00000301030.10	NM_013275.6
NC_000016.10:g.89284211_89284214del	ENST00000301030.10:c.2329_2332del	ENSP00000301030.4:p.Glu777Arg	frameshift_variant	ANKRD11	ENSG00000167522	ENST00000301030.10	NM_013275.6
NC_000016.10:g.89510539T>A	ENST00000268704.7:c.233T>A	ENSP00000268704.3:p.Leu78Ter	stop_gained	SPG7	ENSG00000197912	ENST00000268704.7	
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NC_000016.10:g.89531965_89531993del	ENST00000268704.7:c.1028_1056del	ENSP00000268704.3:p.Pro343Gln	frameshift_variant	SPG7	ENSG00000197912	ENST00000268704.7	
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NC_000016.10:g.89546737C>T	ENST00000268704.7:c.1508C>T	ENSP00000268704.3:p.Ala503Val	missense_variant	SPG7	ENSG00000197912	ENST00000268704.7	
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NC_000016.10:g.89546737C>T	ENST00000268704.7:c.1508C>T	ENSP00000268704.3:p.Ala503Val	missense_variant	SPG7	ENSG00000197912	ENST00000268704.7	
NC_000016.10:g.89546737C>T	ENST00000268704.7:c.1508C>T	ENSP00000268704.3:p.Ala503Val	missense_variant	SPG7	ENSG00000197912	ENST00000268704.7	
NC_000016.10:g.89546737C>T	ENST00000268704.7:c.1508C>T	ENSP00000268704.3:p.Ala503Val	missense_variant	SPG7	ENSG00000197912	ENST00000268704.7	
NC_000016.10:g.89546737C>T	ENST00000268704.7:c.1508C>T	ENSP00000268704.3:p.Ala503Val	missense_variant	SPG7	ENSG00000197912	ENST00000268704.7	
NC_000016.10:g.89550579G>C	ENST00000268704.7:c.1728G>C	ENSP00000268704.3:p.Trp576Cys	missense_variant	SPG7	ENSG00000197912	ENST00000268704.7	
NC_000016.10:g.89553855del	ENST00000268704.7:c.1977del	ENSP00000268704.3:p.Met660Trp	frameshift_variant	SPG7	ENSG00000197912	ENST00000268704.7	
NC_000016.10:g.89554714T>G	ENST00000268704.7:c.2160+151T>G		intron_variant	SPG7	ENSG00000197912	ENST00000268704.7	
NC_000016.10:g.89562922_89562948dup	ENST00000311528.10:c.516_542dup	ENSP00000307889.5:p.Ala180_Se	inframe_insertion	RPL13	ENSG00000167526	ENST00000311528.10	NM_000977.4
NC_000016.10:g.89562954G>A	ENST00000311528.10:c.548G>A	ENSP00000307889.5:p.Arg183His	missense_variant	RPL13	ENSG00000167526	ENST00000311528.10	NM_000977.4
NC_000016.10:g.89562954G>A	ENST00000311528.10:c.548G>A	ENSP00000307889.5:p.Arg183His	missense_variant	RPL13	ENSG00000167526	ENST00000311528.10	NM_000977.4
NC_000016.10:g.89935140C>T	ENST00000315491.12:c.689C>T	ENSP00000320295.7:p.Ser230Leu	missense_variant	TUBB3	ENSG00000258947	ENST00000315491.12	NM_006086.4
NC_000017.11:g.553417G>A	ENST00000291074.10:c.1663C>T	ENSP00000291074.5:p.Arg555Ter	stop_gained	VPS53	ENSG00000141252	ENST00000291074.10	
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NC_000023.11:g.9746073G>A	ENST00000431126.1:c.377C>T	ENSP00000406138.1:p.Pro126Leu	missense_variant	GPR143	ENSG00000101850	ENST00000431126.1	
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NC_000023.11:g.48923703_48923704del	ENST00000156084.8:c.1527_1528del	ENSP00000156084.4:p.Leu510Cys	frameshift_variant	OTUD5	ENSG00000068308	ENST00000156084.8	
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NC_000023.11:g.53254626_53254646delins	ENST00000375365.2:c.670_690delinsT	ENSP00000364514.2:p.Gly224Leu	frameshift_variant	IQSEC2	ENSG00000124313	ENST00000375365.2	
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NC_000023.11:g.53536610C>A	ENST00000262854.11:c.12195G>T	ENSP00000262854.6:p.Trp4065Cy	missense_variant	HUWE1	ENSG00000086758	ENST00000262854.11	NM_031407.7
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NC_000023.11:g.71132894C>G	ENST00000333646.11:c.4345C>G	ENSP00000333125.8:p.Leu1449Va	missense_variant	MED12	ENSG00000184634	ENST00000333646.11	
NC_000023.11:g.71223879C>T	ENST00000361726.7:c.172C>T	ENSP00000354900.6:p.Pro58Ser	missense_variant	GJB1	ENSG00000169562	ENST00000361726.7	NM_000166.6
NC_000023.11:g.71223879C>T	ENST00000361726.7:c.172C>T	ENSP00000354900.6:p.Pro58Ser	missense_variant	GJB1	ENSG00000169562	ENST00000361726.7	NM_000166.6
NC_000023.11:g.71376995C>T			upstream_gene_variant	TAF1	ENSG00000147133	ENST00000276072.9	
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NC_000023.11:g.72464666G>T	ENST00000373568.7:c.803C>A	ENSP00000362669.3:p.Thr268Lys	missense_variant	HDAC8	ENSG00000147099	ENST00000373568.7	
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NC_000023.11:g.108170821G>A	ENST00000334504.12:c.3374C>T	ENSP00000334733.7:p.Pro1125Le	missense_variant	COL4A6	ENSG00000197565	ENST00000334504.12	NM_033641.4
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NC_000023.11:g.132082362C>A	ENST00000298542.9:c.905+1G>T		splice_donor_variant	FRMD7	ENSG00000165694	ENST00000298542.9	NM_194277.3
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NC_000023.11:g.155261209T>C	ENST00000369454.4:c.236A>G	ENSP00000358466.3:p.Tyr79Cys	missense_variant	RAB39B	ENSG00000155961	ENST00000369454.4	NM_171998.4	
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HGVSg CNV

seq[GRCh38] 2p16.3p16.3(48707059_48711185)x1
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seq[GRCh38] del(8)(q22.3q22.3) NC_000008.11:g.102229073_102234572del
seq[GRCh38] del(7)(q36.3)dn
seq[GRCh38] del(1)(p36.33)pat,del(1)(p36.33)x2
Homozygous deletion of approximately 136.5kb on chromosome 15q15.3, carrying the entire STRC gene.
seq[GRCh38] 4q21.21q21.21(78249487_78254789)x0
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NC_000009.12:g.72679942_72722296del
seq[GRCh38] 12p13.33p11.1(1092001_34692000)x3~4
seq[GRCh38] 1p21.1p21.1(101997045_103282337)x1
seq[GRCh38] del(19)(q13.42q13.42) NC_000019.10:g.54123872_(54131713_?)del
NC_000016.10:g.28482327_28483205del
seq[GRCh38] del(1)(p36.22p36.22)pat NC_000001.11:g.9940589_9948917del
seq[GRCh38] t(X;22)(p22.11;q13.33) NC_000022.11:49806740_qterdelins[NC_000023.11:g.pter_22240874inv] NC_000023.11:g.pter_22240877delins[NC_000022.11:g.49806741_qterinv]
seq[GRCh38] 7q11.23q11.23(73268001_74832000)x1
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STRC deletion
seq[GRCh38] del(19)(p13.11p13.11)pat NC_000019.10:g.16974327_16975440del
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seq[GRCh38] 12q14.2q14.2(64463877_64473125)x0
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seq[GRCh38] del(3)(p13p13)dn NC_000003.12:g.70986537_70990562del
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seq[GRCh38] del(1)(p36.33p36.33) NC_00001.11:g.1484701_1519450del
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seq[GRCh38] del(X)(p11.4p11.4)dn NC_000023.11:g. 41609262_41616693del
seq[GRCh38] del(X)(q23q23) NC_000023.11:g.110025622_110209539del
seq[GRCh38] inv(X)(p22.11p22.11)mat NC_000023.11:g.[22035015_22037626del;22037626_22058856inv;22058857_22072045del]
seq[GRCh38] 16q24.1(85891788_86779588x4)dn,16q24.1q24.2(86779589_87030499x3)dn
seq[GRCh38] del(9)(q21.2q21.2) NC_000009.12:g.77242600_77248052del
seq[GRCh38] mos der(18)t(7;18)(q34;q23)dn NC_000018.10:g.75662838_qterdelins[NC_000007.14:g.(?_141230002)_qter]
seq[GRCh38] del(17) NC_00017.11:g.19648751_19677596del
seq[GRCh38] del(X)(p22.2p22.2)dn NC_000023.11:g.103519823_103674293del
seq[GRCh38] inv(15)(q23q26.1)mat NC_000015.10:g.[70833812_88844921inv;88844922_88844930del]
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seq[GRCh38] Xp11.23(49071343_49082968)x0 dn
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seq[GRCh38] del(15) NC_000015.10:g.89206896_89214257del
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seq(4,9)cx dn
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seq[GRCh38] 15q11.2q11.2(22550001_23042000)x1 mat
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Complex chromosomal rearrangement implicating chromosomes 1, 4 and 12
l; ENST00000636147.2(CLN3):c.641-
seq[GRCh38] Xq22.2q22.2(103512832_104448388)x1 dn
seq[GRCh38] del(17)(q11.2q11.2) NC_000017.11:g.31089144_31113701del
seq[GRCh38] del(14)(q32.33q32.33) NC_000014.9:g.104699484_104703736del
seq[GRCh38] del(17)(p12p11.2) NC_000017.11:g.15820295_20638116del
NC_000023.11:g.[?_154412783inv;154412783del]
NC_000016.10:g.29540001_30188000del
NC_000001.11:g.146723001_148071000del
NC_000017.11:g.29500001_29800000del
NC_000006.12:g.168900001_170805979del
NC_000012.12:g.1_1650000del
NC_000016.10:g.15350001_16630000del
NC_000005.10:g.83866712_91209607del
NC_000007.14:g.73120001_74800000dup
NC_000019.10:g.54117731_54121182del
NC_000022.11:g.18947001_21390000del
NC_000008.11:g.76851432_76853236del
NC_00015.10:g.24166672_101751393inv
NC_000001.11:g.146300001_148500000del
NC_000002.12:g.110092811_110230076del
seq[GRCh38] t(15;16) NC_000015.10:g.48449249 NC_000016.10:g.12271054
NC_000023.11:g.154158027_154312265del
NC_000022.11:g.18924718_21111383del
NC_000002.12:g.148499909_148504602del
NC_000017.11:g.44897001_44961000del
NC_000010.11:g.86480001_87140000del
NC_000017.11:g.36340001_38250000del
NC_000012.12:g.115974652_115978220del
NC_000015.10:g.48444001_48454000dup
NC_000001.11:g.23020518_23026884del
NC_000016.10:g.29500001_30205000del
NC_000005.10:g.54880001_58640000del
NC_000007.14:g.4129309_4129369del
NC_000023.11:g.154100001_154300000del
NC_000016.10:g.29540001_30200000del
NC_000006.12:g.157040001_157133000del
seq[GRCh38] inv(11)(q22.1q24.1) NC_000010.11:g.102253109_123049929inv
seq[GRCh38] t(5;18) NC_00005.10:g.94804166 NC_000018.10:g.47562882
NC_000002.12:g.174380001_176940000dup
NC_000001.11:g.146200001_148520000del
NC_000002.12:g.49939001_49980000del
NC_000024.10:g.pter_qteradd
NC_000004.12:g.88043856_88050314del
NC_000010.11:g.92541446_92607801del
seq[GRCh38] inv(X)(p11.22p11.1) NC_000023.11:g.53947212_58520696inv
NC_000020.11:g.58562001_58713000dup
NC_000007.14:g.152220001_152282000del
NC_000022.11:g.26599662_26600310del
NC_000023.11:g.10900001_11120000del
NC_000003.12:g.2085001_9815000del
NC_000006.12:g.64897655_64996334del
NC_000001.11:g.43395480_43405192del
NC_000016.10:g.28485964_28486930del
NC_000023.11:g.153922121_153936508delinsCGTCTC
NC_000023.11:g.48887895_48888770del
NC_000002.12:g.166109001_166190000del
NC_000023.11:g.47971599_47971703del
NC_000016.10:g.15330001_16520000del
NC_000001.11:g.102972381_102975888del
NC_000002.12:g.110450001_112430000dup

NC_000001.11:g.23050038_23093073del
seq[GRCh38] inv(2)(p16.1p16.1) NC_000002.12:g.55672849_60547158inv
NC_000015.10:g.95900001_96600000del
NC_000006.12:g.104794375_104805190del
NC_000018.10:g.62152641_62157701del
NC_000022.11:g.20360001_21130000del
NC_000016.10:g.88980001_90200000dup
NC_000016.10:g.29500001_30200000del
NC_000005.10:g.177992214_178006000del
NC_000015.10:g.22549001_23040000del

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