Chromoso	Region	Type	Reference	Allele	Zygosity
1	2441557	SNV	C	T	Heterozygous
1	7797589	Deletion	A	–	Heterozygous
1	7890069	SNV	C	T	Heterozygous
1	109394866	SNV	T	C	Heterozygous
1	151379728	Insertion	–	T	Heterozygous
1	158908273	SNV	G	C	Heterozygous
1	180023062	Insertion	–	A	Heterozygous
2	73784436	SNV	G	A	Heterozygous
2	95952931	Insertion	–	A	Heterozygous
2	220354194	Insertion	–	C	Heterozygous
3	49395678	MNV	CC	GG	Heterozygous
3	98503874	SNV	G	C	Heterozygous
4	3133499	SNV	C	T	Heterozygous
4	5733312	SNV	C	A	Heterozygous
5	137621817	SNV	C	T	Heterozygous
5	179069957	Insertion	–	AG	Heterozygous
6	31922379	MNV	CT	TC	Heterozygous
6	33147231	Insertion	–	G	Heterozygous
6	161469775	Insertion	–	A	Heterozygous
6	167754563	SNV	C	A	Heterozygous
7	139268736	Insertion	–	G	Heterozygous
8	62212438	SNV	G	A	Heterozygous
8	145005749	SNV	G	A	Heterozygous
9	37746469	SNV	C	G	Heterozygous
10	134010607	Deletion	TGCCGTC	–	Heterozygous
11	1247464	SNV	G	A	Heterozygous
11	74109201	Insertion	–	C	Heterozygous
14	94203633	SNV	C	T	Heterozygous
14	94578113	Deletion	C	–	Heterozygous
15	75132030	SNV	G	A	Heterozygous
16	56368666	Deletion	G	–	Heterozygous
17	7330170	SNV	C	G	Heterozygous
17	7812675	SNV	C	G	Heterozygous
17	40128774	MNV	TC	AA	Heterozygous
17	41610607	Insertion	–	G	Heterozygous
18	24126882	SNV	A	G	Heterozygous
18	24126886	SNV	A	C	Heterozygous
19	1621970	Deletion	C	–	Heterozygous
19	1621970	MNV	CT	TG	Heterozygous
19	2280741	SNV	T	C	Heterozygous
19	15651383	SNV	G	A	Heterozygous
19	17306197	SNV	G	A	Heterozygous
19	21992360	SNV	T	G	Heterozygous
19	38964327	MNV	GC	CG	Heterozygous
19	41349879	SNV	T	C	Heterozygous
19	43429907	SNV	A	G	Heterozygous
19	44771144	SNV	G	A	Heterozygous
19	46318091	SNV	G	A	Heterozygous
19	48807246	Insertion	–	CTGC	Heterozygous
19	52941908	SNV	C	T	Heterozygous
19	53015217	SNV	G	A	Heterozygous
19	55295164	SNV	A	C	Heterozygous
21	34924024	Deletion	C	–	Heterozygous
22	50657144	Insertion	–	TCG	Heterozygous

Coding.region.change
ENST00000505228:c.[*96G>A]; ENST00000378466:c.[1978G>A]; ENST00000502512:c.[*96G>A]; ENST00000435556:c.[1861G>A]
ENST00000303635:c.[3617delA]; ENST00000439411:c.[3617delA]; ENST00000495233:c.[485delA]
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ENST00000367607:c.5167_5168insA
ENST00000423048:c.[*587G>A]; ENST00000264448:c.[10165G>A]; ENST00000409009:c.[10039G>A]
ENST00000542147:c.[1998_1999insA]; ENST00000431567:c.[*1626_*1627insA]; ENST00000317620:c.[2145_2146insA]; ENST00000403131:c.[2145_2146insA]; ENST00000317668:c.[2145_2146insA]
ENST00000312358:c.8454_8455insC
ENST00000419349:c.[33_34delGGinsCC]; ENST00000419783:c.[33_34delGGinsCC]
ENST00000485145:c.[163G>C]; ENST00000483910:c.[421G>C]; ENST00000265261:c.[78-3006G>C]; ENST00000497008:c.[336-3006G>C]; ENST00000486334:c.[421G>C]; ENST00000497621:c.[*554G>C]; ENST00000394162:c.[421G>C]
ENST00000355072:c.2233C>T
ENST00000509451:c.[545C>A]; ENST00000264956:c.[545C>A]; ENST00000382674:c.[545C>A]
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ENST00000512899:c.[595_596insCT]; ENST00000513845:c.[*504_*505insCT]; ENST00000448248:c.[595_596insCT]
ENST00000454913:c.[694_695delAGinsGA]; ENST00000436289:c.[679_680delAGinsGA]; ENST00000441998:c.[679_680delAGinsGA]; ENST00000444811:c.[604_605delAGinsGA]; ENST00000375429:c.[694_695delAGinsGA]; ENST00000375
ENST00000374712:c.[1236_1237insC]; ENST00000374714:c.[1401_1402insC]; ENST00000341947:c.[1479_1480insC]; ENST00000457788:c.[1479_1480insC]; ENST00000374713:c.[1338_1339insC]; ENST00000395197:c.[1299_1300insC]; EN
ENST00000366920:c.[471_472insA]; ENST00000544041:c.[471_472insA]; ENST00000366919:c.[471_472insA]; ENST00000392142:c.[471_472insA]; ENST00000490904:c.[471_472insA]; ENST00000348824:c.[471_472insA]; ENST0000054295
ENST00000515138:c.[1175C>A]; ENST00000239587:c.[1175C>A]
ENST00000406875:c.[2791_2792insC]; ENST00000342645:c.[2491_2492insC]; ENST00000428878:c.[2710_2711insC]
ENST00000518592:c.[-383+11741G>A]; ENST00000325897:c.[52G>A]; ENST00000519846:c.[52G>A]; ENST00000522621:c.[52G>A]
ENST00000436759:c.[2338C>T]; ENST00000357649:c.[2269C>T]; ENST00000354958:c.[2191C>T]; ENST00000354589:c.[2257C>T]; ENST00000527096:c.[2326C>T]; ENST00000356346:c.[2215C>T]; ENST00000322810:c.[2668C>T]; ENS
ENST00000377765:c.[4440C>G]; ENST00000539465:c.[4440C>G]; ENST00000540557:c.[*911-18444G>C]
ENST00000338492:c.[621+2_621+40delTGCCGTGGGGCAGGGCTGCCGTGGGGCAGGCACAGGGGC]; ENST00000368629:c.[390+2_390+40delTGCCGTGGGGCAGGGCTGCCGTGGGGCAGGCACAGGGGC]; ENST00000368627:c.[390+2_3
ENST00000529681:c.[157G>A]; ENST00000447027:c.[157G>A]
ENST00000298198:c.5_6insG
ENST00000393140:c.[313G>A]; ENST00000477603:c.[313G>A]; ENST00000393143:c.[313G>A]; ENST00000316227:c.[313G>A]
ENST00000554448:c.[85delC]; ENST00000557634:c.[85delC]; ENST00000557098:c.[-45+970delC]; ENST00000554800:c.[85delC]; ENST00000556544:c.[85delC]; ENST00000298902:c.[85delC]; ENST00000444961:c.[85delC]; ENST000005
ENST00000568210:c.[*605C>T]; ENST00000568667:c.[754C>T]; ENST00000568273:c.[*528C>T]; ENST00000569437:c.[721C>T]; ENST00000440863:c.[721C>T]; ENST00000566631:c.[*389C>T]
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ENST00000333870:c.[860C>G]; ENST00000574034:c.[*144C>G]
ENST00000358181:c.[5488+19C>G]; ENST00000449744:c.[466+19C>G]; ENST00000380358:c.[5767+19C>G]; ENST00000330494:c.[5590+19C>G]; ENST00000439235:c.[593C>G]; ENST00000573936:c.[547+19C>G]; ENST00000470531:c.
ENST00000457167:c.[1461_1462delGAinsTT]; ENST00000393892:c.[*2832_*2833delTCinsAA]; ENST00000426588:c.[1293_1294delGAinsTT]; ENST00000316603:c.[1293_1294delGAinsTT]; ENST00000587727:c.[144_145delGAinsTT]; ENST
ENST00000393664:c.[492_493insC]; ENST00000545089:c.[384-301_384-300insC]; ENST00000591713:c.[492_493insC]; ENST00000589898:c.[351-3173_351-3172insG]; ENST00000545954:c.[375_376insC]; ENST00000538265:c.[375_376in
ENST00000317932:c.[-15-45668T>C]; ENST00000579973:c.[-15-45668T>C]; ENST00000578973:c.[-206T>C]; ENST00000580059:c.[-206T>C]; ENST00000408011:c.[-16+1973T>C]; ENST00000580638:c.[-16+2811T>C]; ENST00000580
ENST00000579973:c.[-15-45672T>G]; ENST00000580059:c.[-210T>G]; ENST00000578973:c.[-210T>G]; ENST00000580191:c.[10-45672T>G]; ENST00000580638:c.[-16+2807T>G]; ENST00000417602:c.[1615T>G]; ENST00000317932:c.
ENST00000453954:c.[571-1delG]; ENST00000262965:c.[823-1delG]; ENST00000395423:c.[670-1delG]; ENST00000344749:c.[823-1delG]; ENST00000588136:c.[823-1delG]
ENST00000344749:c.[823-2_823-1delAGinsCA]; ENST00000395423:c.[670-2_670-1delAGinsCA]; ENST00000588136:c.[823-2_823-1delAGinsCA]; ENST00000262965:c.[823-2_823-1delAGinsCA]; ENST00000453954:c.[571-2_571-1delAG
ENST00000342063:c.[82+108A>G]; ENST00000590316:c.[190A>G]
ENST00000269703:c.[794G>A]; ENST00000601005:c.[794G>A]
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ENST00000599906:c.[284A>C]; ENST00000595461:c.[461A>C]; ENST00000354959:c.[479A>C]; ENST00000598381:c.[461A>C]; ENST00000594012:c.[461A>C]
ENST00000359596:c.[4076_4077delGCinsCG]; ENST00000355481:c.[4076_4077delGCinsCG]; ENST00000360985:c.[4076_4077delGCinsCG]
ENST00000301141:c.[1307A>G]; ENST00000601627:c.[117+42559T>C]
ENST00000446844:c.[1261T>C]; ENST00000406070:c.[1243+18T>C]
ENST00000592581:c.[268G>A]; ENST00000592844:c.[238+31817C>T]; ENST00000391958:c.[238+30G>A]; ENST00000334152:c.[590+30G>A]; ENST00000590668:c.[238+30G>A]; ENST00000589522:c.[238+30G>A]; ENST00000589799:c.[
ENST00000597055:c.[344C>T]; ENST00000221538:c.[344C>T]
ENST00000315396:c.705_706insGCAG
ENST00000433050:c.[1195C>T]; ENST00000332323:c.[1234C>T]; ENST00000301085:c.[271+3446C>T]; ENST00000432303:c.[142+4574C>T]
ENST00000421239:c.1583G>A
ENST00000541392:c.[35-33825A>C]; ENST00000336077:c.[946A>C]; ENST00000402254:c.[35-33825A>C]; ENST00000434419:c.[946A>C]; ENST00000538269:c.[35-33825A>C]; ENST00000291633:c.[1024A>C]; ENST00000396284:c.[35
ENST00000300278:c.[2487delC]; ENST00000290239:c.[2487delC]; ENST00000356577:c.[2487delC]; ENST00000455528:c.[2487delC]; ENST00000381679:c.[2487delC]; ENST00000381692:c.[244+5339delC]
ENST00000425018:c.[866_867insCGA]; ENST00000248846:c.[4808_4809insCGA]; ENST00000439308:c.[*385_*386insCGA]

Amino.acid.change
ENSP00000367727:p.[Val660Met]; ENSP00000421433:p.[Val621Met]
ENSP00000306522:p.[Ile1207fs]; ENSP00000402561:p.[Ile1207fs]; ENSP00000451720:p.[Ile163fs]
ENSP00000355031:p.[Ser1012Leu]; ENSP00000366755:p.[Ser1021Leu]
ENSP00000359018:p.[Ser141Gly]; ENSP00000359012:p.[Ser141Gly]; ENSP00000359011:p.[Ser141Gly]; ENSP00000432262:p.[Ser141Gly]; ENSP00000436835:p.[Ser141Gly]; ENSP00000433889:p.[Ser141Gly]
ENSP00000354467:p.[Phe753fs]; ENSP00000443547:p.[Phe168fs]; ENSP00000418408:p.[Phe762fs]; ENSP00000431259:p.[Phe744fs]; ENSP00000376484:p.[Phe753fs]; ENSP00000386836:p.[Phe797fs]; ENSP00000432295:p.[Phe206fs]; E
ENSP00000357117:p.[Ala118Pro]; ENSP00000376083:p.[Ala118Pro]; ENSP00000376082:p.[Ala109Pro]; ENSP00000407616:p.[Ala118Pro]; ENSP00000357122:p.[Ala118Pro]; ENSP00000357120:p.[Ala109Pro]
ENSP00000356579:p.His1726fs
ENSP00000264448:p.[Ala3389Thr]; ENSP00000386627:p.[Ala3347Thr]
ENSP00000442542:p.[Pro667fs]; ENSP00000318270:p.[Pro716fs]; ENSP00000385716:p.[Pro716fs]; ENSP00000318520:p.[Pro716fs]
ENSP00000311684:p.Val2819fs
ENSP00000391316:p.[Ala12Pro]; ENSP00000407375:p.[Ala12Pro]
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ENSP00000355887:p.[Asn160fs]; ENSP00000445018:p.[Asn160fs]; ENSP00000355886:p.[Asn160fs]; ENSP00000375986:p.[Asn160fs]; ENSP00000446303:p.[Asn160fs]; ENSP00000297332:p.[Asn160fs]
ENSP00000424130:p.[Ala392Asp]; ENSP00000239587:p.[Ala392Asp]
ENSP00000385571:p.[Ser931fs]; ENSP00000343108:p.[Ser831fs]; ENSP00000413724:p.[Ser904fs]
ENSP00000325506:p.[Gly18Arg]; ENSP00000428402:p.[Gly18Arg]; ENSP00000428986:p.[Gly18Arg]
ENSP00000388180:p.[Arg780Cys]; ENSP00000350277:p.[Arg757Cys]; ENSP00000347044:p.[Arg731Cys]; ENSP00000346602:p.[Arg753Cys]; ENSP00000434583:p.[Arg776Cys]; ENSP00000348702:p.[Arg739Cys]; ENSP00000323856:p.[Arg
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ENSP00000436812:p.[Val53Met]; ENSP00000415793:p.[Val53Met]
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ENSP00000376848:p.[Val105Met]; ENSP00000434370:p.[Val105Met]; ENSP00000376851:p.[Val105Met]; ENSP00000320948:p.[Val105Met]
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ENSP00000466042:p.Met64Val
ENSP00000269703:p.[Arg265Gln]; ENSP00000469866:p.[Arg265Gln]
ENSP00000380444:p.[Ala1321Thr]; ENSP00000472915:p.[Ala1321Thr]; ENSP00000471367:p.[Ala1321Thr]; ENSP00000471457:p.[Ala1321Thr]
ENSP00000469424:p.[Asn95Thr]; ENSP00000469645:p.[Asn154Thr]; ENSP00000347045:p.[Asn160Thr]; ENSP00000470275:p.[Asn154Thr]; ENSP00000469578:p.[Asn154Thr]
ENSP00000352608:p.[Gly1359Ala]; ENSP00000347667:p.[Gly1359Ala]; ENSP00000354254:p.[Gly1359Ala]
ENSP00000301141:p.Lys436Arg
ENSP00000470856:p.Ser421Pro
ENSP00000468154:p.Gly90Arg
ENSP00000472630:p.[Thr115Ile]; ENSP00000221538:p.[Thr115Ile]
ENSP00000318429:p.Pro236fs
ENSP00000391358:p.[Arg399*]; ENSP00000327538:p.[Arg412*]
ENSP00000459216:p.Arg528His
ENSP00000336769:p.[Thr316Pro]; ENSP00000415758:p.[Thr316Pro]; ENSP00000291633:p.[Thr342Pro]
ENSP00000300278:p.[Ser830fs]; ENSP00000290239:p.[Ser830fs]; ENSP00000348984:p.[Ser830fs]; ENSP00000399783:p.[Ser830fs]; ENSP00000371095:p.[Ser830fs]
ENSP00000405979:p.[Leu288_Glu289insAsp]; ENSP00000248846:p.[Leu1602_Glu1603insAsp]

Amino.acid.change.in.longest.transcript	Coding.region.change.in.longest.transcript	Other.variants.within.codon	Non.synonymous	Splice.effect
ENSP00000367727:p.Val660Met	ENST00000378466:c.1978G>A	No	Yes	
ENSP00000306522:p.Ile1207fs	ENST00000303635:c.3617delA	No	Yes	
ENSP00000366755:p.Ser1021Leu	ENST00000377532:c.3062C>T	No	Yes	
ENSP00000359018:p.Ser141Gly	ENST00000370001:c.421A>G	No	Yes	
ENSP00000271715:p.Phe806fs	ENST00000271715:c.2414_2415insA	No	Yes	
ENSP00000357122:p.Ala118Pro	ENST00000368140:c.352G>C	No	Yes	
ENSP00000356579:p.His1726fs	ENST00000367607:c.5167_5168insA	No	Yes	
ENSP00000264448:p.Ala3389Thr	ENST00000264448:c.10165G>A	No	Yes	
ENSP00000318270:p.Pro716fs	ENST00000317620:c.2145_2146insA	No	Yes	
ENSP00000311684:p.Val2819fs	ENST00000312358:c.8454_8455insC	No	Yes	
ENSP00000407375:p.Ala12Pro	ENST00000419783:c.33_34delGGinsCC	No	Yes	
	ENST00000265261:c.78-3006G>C	No	Yes	
ENSP00000347184:p.Pro745Ser	ENST00000355072:c.2233C>T	No	Yes	
ENSP00000372120:p.Thr182Asn	ENST00000382674:c.545C>A	No	Yes	
ENSP00000321656:p.Arg389His	ENST00000323760:c.1166G>A	No	Yes	
ENSP00000404583:p.Glu199fs	ENST00000448248:c.595_596insCT	No	Yes	
ENSP00000364578:p.Arg232Glu	ENST00000375429:c.694_695delAGinsGA	No	Yes	
ENSP00000339915:p.Tyr494fs	ENST00000341947:c.1479_1480insC	No	Yes	
ENSP00000375986:p.Asn160fs	ENST00000392142:c.471_472insA	No	Yes	
ENSP00000424130:p.Ala392Asp	ENST00000515138:c.1175C>A	No	Yes	
ENSP00000385571:p.Ser931fs	ENST00000406875:c.2791_2792insC	No	Yes	
ENSP00000428402:p.Gly18Arg	ENST00000519846:c.52G>A	No	Yes	
ENSP00000323856:p.Arg890Cys	ENST00000322810:c.2668C>T	No	Yes	
ENSP00000444411:p.Asp1480Glu	ENST00000539465:c.4440C>G	No	Yes	
	ENST00000338492:c.621+2_621+40delTGCCGTGGGGCAGGGCTGCCGTGGGGCAGGCACAGGGGC	No	-	Possible splice site disruption
ENSP00000415793:p.Val53Met	ENST00000447027:c.157G>A	No	Yes	
ENSP00000298198:p.Glu3fs	ENST00000298198:c.5_6insG	No	Yes	
ENSP00000376848:p.Val105Met	ENST00000393140:c.313G>A	No	Yes	
ENSP00000451956:p.Leu30fs	ENST00000555744:c.85delC	No	Yes	
ENSP00000400312:p.Arg241*	ENST00000440863:c.721C>T	No	Yes	
ENSP00000262494:p.Ala165fs	ENST00000262494:c.490delG	No	Yes	
ENSP00000328061:p.Pro287Arg	ENST00000333870:c.860C>G	No	Yes	
	ENST00000380358:c.5767+19C>G	No	Yes	
	ENST00000393892:c.*2832_*2833delTCinsAA	No	Yes	
ENSP00000321835:p.Ser165fs	ENST00000319349:c.492_493insC	No	Yes	
ENSP00000408405:p.Val540Ala	ENST00000417602:c.1619T>C	No	Yes	
ENSP00000408405:p.Ser539Ala	ENST00000417602:c.1615T>G	No	Yes	
	ENST00000262965:c.823-1delG	No	-	Possible splice site disruption
	ENST00000262965:c.823-2_823-1delAGinsCA	No	-	Possible splice site disruption
	ENST00000342063:c.82+108A>G	No	Yes	
ENSP00000269703:p.Arg265Gln	ENST00000269703:c.794G>A	No	Yes	
ENSP00000471457:p.Ala1321Thr	ENST00000595618:c.3961G>A	No	Yes	
ENSP00000469578:p.Asn154Thr	ENST00000594012:c.461A>C	No	Yes	
ENSP00000347667:p.Gly1359Ala	ENST00000355481:c.4076_4077delGCinsCG	No	Yes	
ENSP00000301141:p.Lys436Arg	ENST00000301141:c.1307A>G	No	Yes	
ENSP00000470856:p.Ser421Pro	ENST00000446844:c.1261T>C	No	Yes	
ENSP00000468154:p.Gly90Arg	ENST00000592581:c.268G>A	No	Yes	
ENSP00000221538:p.Thr115Ile	ENST00000221538:c.344C>T	No	Yes	
ENSP00000318429:p.Pro236fs	ENST00000315396:c.705_706insGCAG	No	Yes	
ENSP00000327538:p.Arg412*	ENST00000332323:c.1234C>T	No	Yes	
ENSP00000459216:p.Arg528His	ENST00000421239:c.1583G>A	No	Yes	
	ENST00000538269:c.35-33825A>C	No	Yes	
ENSP00000348984:p.Ser830fs	ENST00000356577:c.2487delC	No	Yes	
	ENST00000439308:c.*385_*386insCGA	No	Yes	

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rs755716268	PANK4
	CAMTA1
rs12750400	PER3
rs181722904	AKNAD1
	POGZ
	PYHIN1
	CEP350
rs758464404	ALMS1
	PROM2
	SPEG
	GPX1
	ST3GAL6
	HTT
	EVC
rs537183556	CDC25C
	C5orf60
	NELFE
	COL11A2
	MAP3K4
rs373611883	TTLL2
	HIPK2
rs770762643	CLVS1
	PLEC
	FRMPD1
	DPYSL4
rs763022013	MUC5B
	PGM2L1
	PRIMA1
	IFI27
	ULK3
	GNAO1
	C17orf74
	CHD3
	DNAJC7
	DHX8, ETV4
	KCTD1
	KCTD1
	TCF3
	TCF3
rs775737691	C19orf35
rs767066474	CYP4F22
rs374157748	MYO9B
	ZNF43
	RYR1
	CYP2A6
rs141117526	PSG7
rs750523558	ZNF233
	RSPH6A
	CCDC114
rs199880338	ZNF534
rs572149997	ZNF578
	KIR2DL1
	SON
	TUBGCP6