Table S11. Experimentally validated silent and missense variants affecting normal splicing

Gene	Chromosome	Position (hg38)	Reference allele	Alternate allele	Amino acid change	cDNA change	Splice site	Experiment RNAseq/ RT-PCR/ Mini gene assay/	Reference
ABCA3	16	2319581	C	T	p.K291K	c.G873A	d1	RT-PCR	[1]
ABCA4	1	94005441	C	T	p.K2049K	c.G6147A	d1	minigene	[2]
ABCA4	1	94023386	C	T	p.R1556K	c.G4667A	d1	minigene	[2]
ABCA4	1	94111579	C	T	p.C54Y	c.G161A	a1	minigene	[2]
ABCA4	1	94024955	T	C	p.S1545G	c.A4633G	d2	RT-PCR	[3]
ABCA4	1	94037350	C	T	p.G1203E	c.G3608A	a1	RT-PCR	[3]
ABCA4	1	94031778	C	G	p.Q1376H	c.G4128C	d1	RT-PCR	[3]
ABCC6	16	16154873	C	T	p.Q1347Q	c.G4041A	d1	RT-PCR	[4]
ABCC8	11	17453119	C	T	p.Q392Q	c.G1176A	d1	RT-PCR	[5]
ABCC8	11	17393697	C	T	p.A1537A	c.G4611A	d1	RT-PCR	[5]
AIMP1	4	106327564	G	A	p.V75M	c.G223A	d1	Minigene	[6]
ARID1B	6	157167185	G	T	p.A1009S	c.G3025T	d1	RNAseq	[7]
ARID2	12	45849775	A	T	p.A637A	c.A1911T	d2	RNAseq	[7]
ARPC1B	7	99391253	G	A	p.A261A	c.G783A	d1	cDNA sequencing	[8]
ASS1	9	130466726	T	G	p.V141G	c.T422G	a2	Minigene	[9]
ASXL1	20	32433917	G	A	p.R573R	c.G1719A	d1	RNAseq	[7]
ATM	11	108281168	G	A	p.K1192K	c.G3576A	d1	cDNA sequencing	[10]
ATM	11	108235834	G	C	p.E166Q	c.G496C	d1	RT-PCR	[11]
ATM	11	108281168	G	A	p.K1192K	c.G3576A	d1	RT-PCR	[12]
ATP6V0A4	7	138739540	C	T	p.P524P	c.G1572A	d1	minigene	[13]
ATP6V1B1	2	70959018	G	T	p.G123V	c.G368T	a1	minigene	[13]
ATP6V1B1	2	70959020	C	T	p.R124W	c.C370T	a3	minigene	[13]
ATP7B	13	51944109	C	T	p.E1081E	c.G3243A	d1	minigene	[14]
ATP7B	13	51946284	C	A	p.K942N	c.G2826T	d1	minigene	[14]
BAP1	3	52402604	T	A	p.E685V	c.A2054T	d3	RT-PCR	[15]
BBS1	11	66515586	G	A	p.R160Q	c.G479A	d1	RT-PCR	[16]
BCAP31	X	153723153	C	T	p.R31K	c.G92A	d1	RT-PCR	[17]

BLNK	10	96223826	C	T	p.E175E	c.G525A	d1	RT-PCR	[18]
BRAT1	7	2540979	C	T	p.T465T	c.G1395A	d1	RT-PCR	[19]
BRCA1	17	43047702	C	G	p.G1803A	c.G5408C	a2	mini-gene	[20]
BRCA1	17	43051063	C	T	p.D1778N	c.G5332A	d1	mini-gene	[20]
BRCA1	17	43051063	C	A	p.D1778Y	c.G5332T	d1	mini-gene	[20]
BRCA1	17	43067608	C	G	p.D1692H	c.G5074C	d1	mini-gene	[20]
BRCA1	17	43067608	C	T	p.D1692N	c.G5074A	d1	mini-gene	[20]
BRCA1	17	43067608	C	A	p.D1692Y	c.G5074T	d1	mini-gene	[20]
BRCA1	17	43067609	T	G	p.T1691T	c.A5073C	d2	RNAseq	[7]
BRCA1	17	43067694	A	T	p.M1663K	c.T4988A	a2	mini-gene	[20]
BRCA1	17	43067695	T	A	p.M1663L	c.A4987T	a1	mini-gene	[20]
BRCA1	17	43067610	G	C	p.T1691R	c.C5072G	d3	RT-PCR	[21]
BRCA1	17	43067610	G	T	p.T1691K	c.C5072A	d3	RT-PCR	[21]
BRCA1	17	43047703	C	A	p.G1803C	c.G5407T	a1	RT-PCR	[22]
BRCA1	17	43076488	C	T	p.R1495K	c.G4484A	d1	RT-PCR	[23]
BRCA1	17	43074331	C	T	p.E1559K	c.G4675A	d1	RT-PCR	[23]
BRCA2	13	32316527	G	A	p.D23N	c.G67A	d1	minigene	[24]
BRCA2	13	32326282	G	A	p.K172K	c.G516A	d1	minigene	[25]
BRCA2	13	32357929	G	C	p.R2602T	c.G7805C	d1	minigene	[26]
BRCA2	13	32363533	G	A	p.K2777K	c.G8331A	d1	minigene	[27]
BRCA2	13	32379913	G	A	p.P3039P	c.G9117A	d1	cDNA sequencing (cBROCA sequencing)	[28]
BRCA2	13	32329444	C	T	p.V211V	c.C633T	a2	RT-PCR	[21]
BRCA2	13	32329491	C	T	p.A227V	c.C680T	d2	RT-PCR	[29]
BRCA2	13	32346895	C	T	p.R2336C	c.C7006T	d2	RT-PCR	[21]
BRCA2	13	32379912	C	T	p.P3039L	c.C9116T	d2	RT-PCR	[21]
BRCA2	13	32344651	A	T	p.D2312V	c.A6935T	d3	RT-PCR	[21]
BRCA2	13	32356609	G	A	p.Q2539Q	c.G7617A	d1	RT-PCR	[21]
BRCA2	13	32325184	G	T	p.S142I	c.G425T	d1	RT-PCR	[30]
CATSPERG	19	38365061	G	A	p.V853M	c.G2557A	a1	MaPSY	[31]
CCDC129	7	31569809	G	C	p.R113T	c.G338C	d1	MaPSY	[31]
CDH23	10	71790413	G	A	p.G2017S	c.G6049A	d1	Minigene	[32]
CDKN2A	9	21974679	T	G	p.Q50P	c.A149C	d2	RT-PCR	[33]
CFTR	7	117531113	A	T	p.K163M	c.A488T	d2	Minigene	[34]

CFTR	7	117542016	G	T	p.D373Y	c.G1117T	a1	Minigene	[34]
CFTR	7	117542108	G	T	p.E403D	c.G1209T	d1	Minigene	[34]
CFTR	7	117611808	G	C	p.G1123R	c.G3367C	d1	Minigene	[34]
CFTR	7	117530899	G	A	p.E92K	c.G274A	a1	RT-PCR	[35]
CFTR	7	117606753	G	A	p.Q996Q	c.G2988A	d1	Minigene	[36]
CFTR	7	117642593	G	C	p.Q1291H	c.G3873C	d1	RT-PCR	[35]
CFTR	7	117627770	G	C	p.R1239S	c.G3717C	d1	RT-PCR	[35]
CFTR	7	117603782	G	C	p.G970R	c.G2908C	d1	RT-PCR	[35],[37], [38]
CFTR	7	117603782	G	A	p.G970S	c.G2908A	d1	RT-PCR	[35]
CHD7	8	60838076	G	T	p.V1452L	c.G4354T	a1	minigene	[39]
CLCN5	X	50070030	G	A	p.E35E	c.G105A	d1	Minigene	[40]
CLCNKB	1	16045686	G	A	p.A77T	c.G229A	d1	minigene	[41]
CLCNKB	1	16045686	G	C	p.A77P	c.G229C	d1	minigene	[41]
CLDN16	3	190408314	G	A	p.G198D	c.G593A	a1	Minigene	[42]
CLDN16	3	190408314	G	C	p.G198A	c.G593C	a1	Minigene	[42]
CLDN19	1	42738421	C	A	p.G130C	c.G388T	d1	Minigene	[43]
CNGB3	8	86739656	T	C	p.Q70Q	c.A210G	d2	Minigene	[44]
CNGB3	8	86668019	C	G	p.D215H	c.G643C	d1	Minigene	[44]
CNGB3	8	86647802	T	C	p.K330R	c.A989G	d2	Minigene	[44]
CNGB3	8	86644622	C	T	p.R352K	c.G1055A	d1	Minigene	[44]
CNGB3	8	86604093	C	G	p.S594T	c.G1781C	d1	Minigene	[44]
CNGB3	8	86578689	C	G	p.Q701H	c.G2103C	d1	Minigene	[44]
CNNM4	2	96797646	A	G	p.T560T	c.A1680G	d2	RNAseq	[7]
COCH	14	30879485	G	T	p.G211C	c.G631T	d1	Minigene	[45]
COL17A1	10	104060121	G	A	p.A380V	c.C1139T	d3	Minigene	[46]
COL17A1	10	104053920	C	T	p.G612R	c.G1834A	d1	Minigene	[46]
COL1A2	7	94408823	G	A	p.K264K	c.G792A	d1	minigene	[47]
COL1A2	7	94422957	G	A	p.G802S	c.G2404A	a1	minigene	[47]
COL27A1	9	114211026	G	A	p.P789P	c.G2367A	d1	RT-PCR	[48]
COL4A3	2	227253638	G	A	p.T255T	c.G765A	d1	Minigene, RT-PCR	[49, 50]
COL4A4	2	227103144	C	T	p.K290K	c.G870A	d1	mRNA Analysis, minigene	[51]
COL4A4	2	227108581	C	T	p.P245P	c.G735A	d1	mRNA Analysis, minigene	[51]

COL4A5	X	108586747	G	A	p.G389R	c.G1165A	d1	Minigene	[52]
COL4A5	X	108591644	G	A	p.G475S	c.G1423A	d1	Minigene	[52]
COL4A5	X	108598870	G	T	p.G650C	c.G1948T	d1	Minigene	[52]
COL4A5	X	108598870	G	A	p.G650S	c.G1948A	d1	Minigene	[52]
COL4A5	X	108601988	A	G	p.K715K	c.A2145G	d2	Minigene	[53]
COL4A5	X	108603061	G	T	p.K748N	c.G2244T	d1	Minigene	[52]
COL4A5	X	108606891	A	T	p.K798N	c.A2394T	d2	Minigene	[53]
COL4A5	X	108606891	A	G	p.K798K	c.A2394G	d2	Minigene	[53]
COL4A5	X	108615024	G	A	p.G837S	c.G2509A	d1	Minigene	[52]
COL4A5	X	108620426	G	C	p.G893R	c.G2677C	d1	Minigene	[52]
COL4A5	X	108620426	G	A	p.G893S	c.G2677A	d1	Minigene	[52]
COL4A5	X	108621892	G	C	p.G923R	c.G2767C	d1	Minigene	[52]
COL4A5	X	108655457	G	A	p.G1125R	c.G3373A	d1	Minigene	[52]
COL4A5	X	108668504	G	A	p.G1264R	c.G3790A	d1	Minigene	[52]
COL4A5	X	108677633	G	C	p.Q1308H	c.G3924C	d1	Minigene	[52]
COL4A5	X	108680751	G	A	p.G1333S	c.G3997A	d1	Minigene	[52]
COL4A5	X	108680956	G	C	p.G1357R	c.G4069C	d1	Minigene	[52]
COL4A5	X	108680956	G	A	p.G1357S	c.G4069A	d1	Minigene	[52]
COL4A5	X	108692925	G	A	p.R1563Q	c.G4688A	d1	Minigene	[52]
COL4A5	X	108695437	C	T	p.F1658F	c.C4974T	d3	Minigene	[53]
COL4A5	X	108695438	A	G	p.S1659G	c.A4975G	d2	Minigene	[53]
COL4A5	X	108695439	G	A	p.S1659N	c.G4976A	d1	Minigene	[52]
COL4A5	X	108621803	G	C	p.G893A	c.G2678C	a1	Minigene	[54]
COL4A5	X	108624236	G	A	p.G973D	c.G2918A	a1	Minigene	[54]
COL5A2	2	189062865	C	T	p.P659P	c.G1977A	d1	RT-PCR	[55]
CRB1	1	197434706	G	A	p.C948Y	c.G2843A	a1	cDNA sequencing	[56]
CYP27B1	12	57765211	C	T	p.G197D	c.G590A	a1	minigene	[57]
CYP27B1	12	57765211	C	T	p.G197D	c.G590A	a1	RT-PCR	[58]
DES	2	219420346	G	C	p.E245D	c.G735C	d1	RT-PCR	[59]
DNMT3A	2	25243899	T	A	p.T645T	c.A1935T	d2	RNAseq	[7]
DSPP	4	87612107	T	C	p.V18V	c.T54C	a3	Minigene	[60]
DSPP	4	87612106	T	A	p.V18D	c.T53A	a2	Minigene	[60]
DSPP	4	87610957	C	A	p.P17T	c.C49A	d3	Minigene	[60]
DSPP	4	87612106	T	G	p.V18G	c.T53G	a2	Minigene	[60]
DSPP	4	87612106	T	C	p.V18A	c.T53C	a2	Minigene	[60]

DSPP	4	87612107	T	A	p.V18V	c.T54A	a3	Minigene	[60]
DSPP	4	87610957	C	T	p.P17S	c.C49T	d3	Minigene	[60]
DYSF	2	71513242	G	A	p.G155R	c.G463A	a3	minigene	[61]
EFTUD2	17	44853390	C	T	p.A823T	c.G2467A	a1	minigene	[62]
EFTUD2	17	44859905	C	G	p.K620N	c.G1860C	d1	minigene	[62]
EFTUD2	17	44859905	C	A	p.K620N	c.G1860T	d1	minigene	[62]
EFTUD2	17	44867807	C	G	p.Q383H	c.G1149C	d1	minigene	[62]
EFTUD2	17	44879556	C	A	p.G234G	c.G702T	d1	cDNA sequencing	[63]
EP300	22	41147946	G	A	p.P747P	c.G2241A	d1	RNAseq	[7]
EPG5	18	45954395	T	C	p.Q336R	c.A1007G	d2	RT-PCR	[64]
EYA4	6	133481599	G	T	p.E369D	c.G1107T	d1	Minigene	[65]
EZH2	7	148826471	T	C	p. H297R	c.A890G	d3	minigene	[66]
EZH2	7	148826631	A	C	p.Y244D	c.T730G	a2	minigene	[66]
F5	1	169523904	C	T	p.G1930D	c.G5789A	a1	Minigene	[67]
F8	X	154987305	C	T	p.G201E	c.G602A	a1	Minigene	[68]
F8	X	154987238	T	A	p.E223D	c.A669T	d2	Minigene	[68]
F8	X	154956957	C	T	p.Q584Q	c.G1752A	d1	Minigene	[69]
F8	X	154987237	C	T	p.G224R	c.G670A	d1	Minigene	[69]
F8	X	154987237	C	A	p.G224W	c.G670T	d1	Minigene	[68]
F8	X	154987238	T	C	p.E223E	c.A669G	d2	Minigene	[68]
F8	X	154969331	C	T	p.D337N	c.G1009A	d1	Minigene	[69]
F8	X	154899866	C	T	p.K2091K	c.G6273A	d1	Minigene	[70]
F9	X	139530851	A	G	p.T29T	c.A87G	d2	RT-PCR	[71]
FAH	15	80173143	A	G	p.Q279R	c.A836G	d2	Minigene	[72]
FANCL	2	58160108	C	T	p.K364K	c.G1092A	d1	RT-PCR	[73]
FBN1	15	48445504	T	C	p.D1930G	c.A5789G	a1	RT-PCR	[22]
FBN1	15	48481656	T	C	p.T1321T	c.A3963G	d2	RT-PCR	[22]
FBN1	15	48513549	C	T	p.D530N	c.G1588A	d1	RT-PCR	[22]
FBN1	15	48485374	C	T	p.D1238N	c.G3712A	d1	RT-PCR	[22]
FECH	18	57571392	C	G	p.A155P	c.G463C	d1	cDNA sequencing	[74]
FLAD1	1	154990526	C	T	p.L518L	c.C1552T	d3	RNAseq	[7]
FMR1	X	147932595	G	A	p.E267E	c.G801A	d1	RT-PCR	[75]
GAA	17	80105132	G	T	p.T182T	c.G546T	d1	RT-PCR	[76]
GALT	9	34648762	G	A	p.E230K	c.G688A	a1	RT-PCR	[77]
GCK	7	44146463	C	T	p.S340N	c.G1019A	d1	minigene	[78]

GCK	7	44146463	C	G	p.S340T	c.G1019C	d1	minigene	[78]
GCK	7	44149760	C	T	p.G227S	c.G679A	d1	minigene	[78]
GCK	7	44149760	C	G	p.G227R	c.G679C	d1	minigene	[78]
GCK	7	44149969	C	A	p.G193G	c.G579T	d1	minigene	[78]
GCK	7	44150064	C	T	p.G162S	c.G484A	a1	minigene	[78]
GCK	7	44153301	C	T	p.E70K	c.G208A	d1	minigene	[78]
GCK	7	44153301	C	G	p.E70Q	c.G208C	d1	minigene	[78]
GCK	7	44152424	T	G	p.E69D	c.A207C	a2	Minigene	[79]
GMPR	6	16274496	G	C	p.G183R	c.G547C	d1	RT-PCR	[80]
GSAP	7	77377287	T	G	p.K227T	c.A680C	d2	MaPSY	[31]
HFM1	1	91276984	C	T	p.C1157Y	c.G3470A	d3	Minigene	[81]
HNF1A	12	120997665	G	A	p.A501T	c.G1501A	d1	minigene	[78]
HNF1A	12	120999389	G	A	p.Q541Q	c.G1623A	d1	minigene	[78]
HNF4A	20	44413800	G	A	p.Q142Q	c.G426A	d1	minigene	[78]
HNF4A	20	44424254	G	C	p.G355R	c.G1063C	d1	minigene	[78]
HSD17B3	9	96240907	C	T	p.V225M	c.G673A	a1	RT-PCR	[82]
IDUA	4	1004111	A	T	p.P609P	c.A1827T	d2	MaPSY	[31]
IFT74	9	26978263	G	A	p.G86S	c.G256A	d1	RT-PCR	[83]
IFT74	9	26978263	G	A	p.G86S	c.G256A	d1	RT-PCR	[84]
IL2RG	X	71108599	C	T	p.R285Q	c.G868A	d1	RT-PCR	[85]
IRF2	4	184418533	T	C	p.K121K	c.A363G	d2	RNAseq	[7]
JAG1	20	10647960	C	G	p.V574L	c.G1720C	d1	cDNA sequencing	[86]
JAK3	19	17835924	C	A	p.L638L	c.G1914T	d1	minigene	[87]
KIAA0586	14	58458545	G	A	p.Q605Q	c.G1815A	d1	RT-PCR	[88]
KIF11	10	92649986	G	A	p.P974P	c.G2922A	d1	minigene	[89]
KMT2C	7	152207301	T	C	p.P1280P	c.A3840G	d2	RNAseq	[7]
LAMB3	1	209633070	C	T	p.E210K	c.G628A	d1	RT-PCR	[90]
LCA5	6	79492551	C	T	p.A319T	c.G955A	d1	cDNA sequence	[91]
LMNA	1	156135312	G	C	p.Q312H	c.G936C	d1	minigene	[92]
LMNA	1	156135312	G	C	p.Q312H	c.G936C	d1	Minigene	[93]
LPIN2	18	2929065	C	T	p.R517H	c. G1550A	d1	RT-PCR	[94]
LRP5	11	68403483	G	T	p.V529L	c.G1585T	a1	Minigene	[21]
LRRK1	15	100973967	G	A	p.K87K	c.G261A	d1	RT-PCR	[95]
MEIOB	16	1839255	C	T	p.T406T	c.G1218A	d1	Minigene	[96]
MFSD8	4	127921524	C	T	p.Q450Q	c.G1350A	d1	RNAseq	[7]

MLH1	3	37042331	G	A	p.S577S	c.G1731A	d1	RT-PCR	[97, 98]
MLH1	3	37049017	G	A	p.Q701Q	c.G2103A	d1	minigene	[99]
MLH1	3	37042331	G	A	p.S577S	c.G1731A	d1	RT-PCR	[100]
MYH9	22	36295505	C	G	p.R1162T	c.G3485C	d1	RT-PCR	[101]
MYO7A	11	77181589	G	A	p.E968E	c.G2904A	d1	minigene	[102]
MYSM1	1	58689038	C	T	p.L133L	c.G399A	d1	RT-PCR	[103]
NCF2	1	183567204	C	G	p.Q285H	c.G855C	d1	RT-PCR	[104]
NCKAP1L	12	54532250	G	C	p.K954N	c.G2862C	d1	MaPSY	[31]
NDUFS2	1	161212480	G	A	p.K372K	c.G1116A	d1	RNAseq	[7]
NF1	17	31258501	A	G	p.K1423R	c.A4268G	d2	cDNA sequencing	[105]
NF1	17	31327839	G	A	p.R1849Q	c.G5546A	d1	cDNA sequencing	[106]
NIPBL	5	37020877	G	A	p.Q1776Q	c.G5328A	d1	RT-PCR	[107]
NPC1	18	23539813	C	T	p.N931N	c.C2793T	d3	Minigene	[108]
NR5A1	9	124503079	C	A	p.A82S	c.G244T	d1	Minigene	[109]
OTC	X	38381429	G	A	p.R129H	c.G386A	d1	Minigene	[110]
P2RX1	17	3898009	C	T	p.A378A	c.G1134A	d1	MaPSY	[31]
PALB2	16	23624093	A	G	p.V917A	c.T2750C	a2	Minigene	[111]
PALB2	16	23607866	G	A	p.G1116G	c.C3348T	d3	Minigene	[111]
PALB2	16	23624009	C	G	p.R945T	c.G2834C	d1	Minigene	[111]
PALB2	16	23629641	T	G	p.Q838P	c.A2513C	d2	Minigene	[111]
PALB2	16	23641111	T	C	p.K16R	c.A47G	d2	Minigene	[111]
PALB2	16	23641110	C	T	p.K16K	c.G48A	d1	Minigene	[111]
PATL2	15	44669563	C	A	p.D293Y	c.G877T	a1	RT-PCR	[112]
PAX6	11	31802705	T	G	p.Q47P	c.A140C	d2	Minigene	[113]
PAX6	11	31802704	C	T	p.Q47Q	c.G141A	d1	Minigene	[113]
PAX6	11	31794631	T	C	p.K227K	c.A681G	d2	Minigene	[113]
PAX6	11	31794630	C	T	p.E228K	c.G682A	d1	Minigene	[113]
PAX6	11	31794033	T	C	p.Q255R	c.A764G	d2	Minigene	[113]
PAX6	11	31794032	C	A	p.Q255H	c.G765T	d1	Minigene	[113]
PAX6	11	31794032	C	G	p.Q255H	c.G765C	d1	Minigene	[113]
PAX6	11	31790710	C	T	p.G395R	c.G1183A	d1	Minigene	[113]
PAX6	11	31802705	T	C	p.Q47R	c.A140G	d2	Minigene	[114]
PGK1	X	78125051	G	A	p.G372S	c.G1114A	d1	RT-PCR	[115]
PHEX	X	22212958	G	C	p.R567P	c.G1700C	d1	minigene	[116]
PHF21A	11	45948886	C	T	p.G429S	c.G1285A	d1	Minigene	[117]

PKD1	16	2092954	C	T	p.R3719Q	c.G11156A	d1	Minigene	[118]
PKD2	4	88052158	G	A	p.K572K	c.G1716A	d1	RT-PCR	[119]
PLG	6	160737006	A	C	p.R601R	c.A1801C	d2	RNAseq	[7]
PNKP	19	49867054	C	G	p.V51L	c.G151C	d1	RT-PCR	[120]
POLR3A	10	77984205	C	T	p.E1112E	c.G3336A	d1	RT-PCR	[121]
POMT2	14	77279823	C	G	p.G631R	c.G1891C	d1	Minigene	[122]
POT1	7	124825252	C	T	p.D598N	c.G1792A	d1	RT-PCR	[123]
POU1F1	3	87273347	C	T	p.G98S	c.G292A	d1	Minigene	[124]
POU1F1	3	87273347	C	G	p.G98R	c.G292C	d1	Minigene	[124]
POU1F1	3	87273347	C	A	p.G98C	c.G292T	d1	Minigene	[124]
POU1F1	3	87273348	T	G	p.A97A	c.A291C	d2	Minigene	[124]
POU1F1	3	87273348	T	C	p.A97A	c.A291G	d2	Minigene	[124]
POU1F1	3	87273348	T	A	p.A97A	c.A291T	d2	Minigene	[124]
POU1F1	3	87273349	G	A	p.A97V	c.C290T	d3	Minigene	[124]
POU1F1	3	87273495	G	C	p.V48V	c.C144G	a2	Minigene	[124]
POU1F1	3	87273496	A	T	p.V48D	c.T143A	a1	Minigene	[124]
POU1F1	3	87273496	A	G	p.V48A	c.T143C	a1	Minigene	[124]
POU1F1	3	87273496	A	C	p.V48G	c.T143G	a1	Minigene	[124]
PPOX	1	161169660	G	T	p.V270L	c.G808T	a1	RT-PCR	[125]
PPP6C	9	125171085	C	T	p.Q57Q	c.G171A	d1	RNAseq	[7]
PRKDC	8	47778459	C	T	p.Q3951Q	c.G11853A	d1	RNAseq	[7]
PRKDC	8	47828168	C	T	p.Q2859Q	c.G8577A	d1	RNAseq	[7]
PTEN	10	87961118	G	A	p.K342K	c.G1026A	d1	RNAseq	[7]
RAD50	5	132604046	G	A	p.V842I	c.G2524A	d1	RT-PCR	[126]
RAD51B	14	68411527	G	A	p.Q319Q	c.G957A	d1	RNAseq	[7]
RAD51D	17	35119532	C	T	p.V28M	c.G82A	d1	RT-PCR	[127]
RGS7	1	240801455	C	T	p.V471V	c.G1413A	d1	MaPSY	[31]
RHD	1	25301521	C	T	p.G212G	c.C636T	a2	Minigene	[128]
RHD	1	25306729	T	C	p.I358T	c.T1073C	d1	Minigene	[128]
RHD	1	25306597	G	T	p.G314V	c.G941T	a2	Minigene	[128]
RHD	1	25284574	T	C	p.V50V	c.T150C	a2	Minigene	[128]
RHD	1	25303458	C	T	p.P313L	c.C938T	d2	Minigene	[128]
RHD	1	25290790	A	G	p.N162S	c.A485G	d2	Minigene	[128]
RHD	1	25301685	A	T	p.K267M	c.A800T	d2	Minigene	[128]

RHD	1	25321889	G	A	p.G385D	c.G1154A	a1	Minigene	[129]
RHD	1	25321889	G	C	p.G385A	c.G1154C	a1	Minigene	[129]
RHD	1	25321889	G	T	p.G385V	c.G1154T	a1	Minigene	[129]
RPE65	1	68431282	C	A	p.R446S	c.G1338T	d1	Minigene	[130]
RPGR	X	38323399	C	T	p.G52R	c.G154A	d1	RT-PCR	[131]
SBF2	11	10029765	C	T	p.Q171Q	c.G513A	d1	RNAseq	[7]
SCN1A	2	165994146	C	T	p.G1618S	c.G4852A	d1	midigene	[132]
SCN1A	2	165994414	G	C	p.N1528K	c.C4584G	a3	Minigene	[124]
SCN1A	2	165998038	C	T	p.K1492K	c.G4476A	d1	Minigene	[124]
SCN1A	2	165999776	C	A	p.A1429S	c.G4285T	a1	Minigene	[124]
SCN1A	2	166041231	C	T	p.L805L	c.G2415A	d1	Minigene	[124]
SCN1A	2	166045043	C	A	p.Q554H	c.G1662T	d1	Minigene	[124]
SCN1A	2	166045043	C	T	p.Q554Q	c.G1662A	d1	Minigene	[124]
SCN1A	2	166045044	T	C	p.Q554R	c.A1661G	d2	Minigene	[124]
SGCA	17	50167487	G	A	p.A53T	c.G157A	d1	RT-PCR	[133]
SLC12A3	16	56870096	G	T	p.G201V	c.G602T	a1	minigene	[134]
SLC12A3	16	56870096	G	A	p.G201D	c.G602A	a1	minigene	[134]
SLC12A3	16	56880253	G	A	p.A523T	c.G1567A	d1	minigene	[134]
SLC12A3	16	56885364	G	A	p.R642H	c.G1925A	d1	minigene	[134]
SLC12A3	16	56893054	G	C	p.G850R	c.G2548C	d1	minigene	[134]
SLC12A3	16	56894531	G	C	p.G850A	c.G2549C	d1	minigene	[134]
SLC40A1	2	189563584	C	T	p.G468S	c.G1402A	a3	minigene	[135]
SLC40A1	2	189572846	G	A	p.L129L	c.C387T	d1	Minigene	[21]
SLC5A2	16	31485730	C	T	p.A102V	c.C305T	a2	Minigene	[136]
SLC5A2	16	31488038	G	C	p.V296L	c.G886C	a1	Minigene	[136]
SLC5A2	16	31488490	G	A	p.G377S	c.G1129A	d1	Minigene	[136]
SMN1	5	70076521	G	C	p.G279R	c.G835C	a1	Minigene	[137]
SPINK1	5	147829599	C	T	p.E29E	c.G87A	d1	FLGSA	[138]
SPINK1	5	147829599	C	G	p.E29D	c.G87C	d1	FLGSA	[138]
SPINK1	5	147829599	C	A	p.E29D	c.G87T	d1	FLGSA	[138]
SPINK1	5	147829600	T	C	p.E29G	c.A86G	d2	FLGSA	[138]
SPINK1	5	147829600	T	A	p.E29V	c.A86T	d2	FLGSA	[138]
SPINK1	5	147829630	C	G	p.G19A	c.G56C	a1	FLGSA	[138]
SPINK1	5	147829630	C	A	p.G19V	c.G56T	a1	FLGSA	[138]
SPINK1	5	147831523	C	T	p.G19S	c.G55A	d1	FLGSA	[138]

SPINK1	5	147831523	C	G	p.G19R	c.G55C	d1	FLGSA	[138]
SPINK1	5	147831523	C	A	p.G19C	c.G55T	d1	FLGSA	[138]
SYNGAP1	6	33432252	G	A	p.S129S	c.G387A	d1	MaPSY	[31]
TCF4	18	55261466	G	A	p.S330S	c.G990A	d1	MaPSY	[31]
TCIRG1	11	68043497	G	A	p.T210T	c.G630A	d1	Minigene	[139]
TMPRSS15	21	18326432	C	T	p.E641K	c.G1921A	d1	Minigene	[140]
TNRC6B	22	40273600	G	A	p.K1047K	c.G3141A	d1	Minigene	[141]
TNRC6C	17	78071165	G	A	p.P953P	c.G2859A	d1	MaPSY	[31]
TP53	17	7673535	C	T	p.Q331Q	c.G993A	d1	minigene	[142, 143]
TP53	17	7674181	C	G	p.S261T	c.G782C	d1	RNAseq	[7]
TP53	17	7674859	C	T	p.E224E	c.G672A	d1	minigene	[142, 144] [145]
TP53	17	7674859	C	A	p.E224D	c.G672T	d1	RNAseq	[7, 145]
TP53	17	7674970	A	T	p.G187G	c.T561A	a2	minigene	[142]
TP53	17	7674970	A	G	p.G187G	c.T561C	a2	minigene	[142]
TP53	17	7674970	A	C	p.G187G	c.T561G	a2	minigene	[142, 144]
TP53	17	7675053	C	G	p.G187R	c.G559C	d1	RNAseq	[7]
TP53	17	7675994	C	T	p.T125T	c.G375A	d1	minigene	[143, 146]
TP53	17	7675994	C	G	p.T125T	c.G375C	d1	minigene	[144, 146]
TP53	17	7675994	C	A	p.T125T	c.G375T	d1	minigene	[142, 144, 146]
TP53	17	7675995	G	A	p.T125M	c.C374T	d2	Minigene	[146]
TP53	17	7675995	G	C	p.T125R	c.C374G	d2	Minigene	[146]
TP53	17	7675996	T	C	p.T125A	c.A373G	d3	Minigene	[146]
TP53	17	7675996	T	G	p.T125P	c.A373C	d3	Minigene	[146, 147]
TREM2	6	41161263	C	T	p.D131N	c.G391A	d1	Minigene	[148]
TRMU	22	46355587	A	G	p.L339L	c.A1017G	d2	RNAseq	[7]
TTC8	14	88872452	G	C	p.Q449H	c.G1347C	d1	cDNA sequencing	[149]

U2AF2	19	55662618	G	T	p.E201D	c.G603T	d1	RT-PCR	[150]
UPF3B	X	119841735	C	A	p.Q208H	c.G624T	d1	RT-PCR	[151]
VWF	12	5976111	C	T	p.S2479S	c.G7437A	d1	cDNA sequencing	[152]
VWF	12	6072331	C	T	p.C370Y	c.G1109A	d1	cDNA sequencing	[152]
ZFN780B	19	40047375	C	T	p.D78N	c.G232A	d1	MaPSY	[31]

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