disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Congenital myasthenic syndrome	AGRN	5179GtoT | V1727F	N/A	1	986143	986143
Myocardial infarction, protection against, assc	TNFRSF4	634plus25CtoT	rs2298212	1	1147297	1147297
Zellweger syndrome	PEX10	600plus1GtoA	N/A	1	2339890	2339890
Zellweger syndrome	PEX10	13_28delGCCGCCAGCCCCCCCGGinsCCGCCAGCACCTGCGCCGCC | 13_28del16ins20 | 13_28delins	N/A	1	2343914	2343929
Zellweger syndrome	PEX10	4delG | 4del1 | 4del	rs62636524	1	2343938	2343938
Zellweger syndrome	PEX10	3GtoA | M1I	N/A	1	2343939	2343939
Ataxia	PEX10	2TtoC | M1T	N/A	1	2343940	2343940
Cardiovascular malformations	NPHP4	3706GtoA | V1236M	N/A	1	5925272	5925272
Nephronophthisis 4	NPHP4	3574CtoT | R1192W	rs139022622	1	5926503	5926503
Nephronophthisis	NPHP4	3403delC | 3403del1 | 3403del	N/A	1	5927869	5927869
Nephronophthisis 4	NPHP4	2836AtoG | T946A	N/A	1	5935142	5935142
Nephronophthisis 4	NPHP4	2203CtoT | R735W	rs191913664	1	5951029	5951029
Nephronophthisis 4	NPHP4	2144minus1GtoC	N/A	1	5951089	5951089
Nephronophthisis 4	NPHP4	1334_1335delTCinsAA | 1334_1335del2ins2 | 1334_1335delins	N/A	1	5987815	5987816
Immunodeficiency, primary B-cell	PIK3CD	3061GtoA | E1021K	N/A	1	9787030	9787030
Charcot-Marie-Tooth disease 2a	MFN2	892GtoA | G298R	rs41278630	1	12061533	12061533
Pancreatitis, chronic	CTRC	494minus10CtoT	N/A	1	15771091	15771091
Parkinsonism with dementia	ATP13A2	3057delC | 3057del1 | 3057del	N/A	1	17313567	17313567
Parkinson disease	ATP13A2	3040GtoA | G1014S	N/A	1	17313584	17313584
Parkinson disease	ATP13A2	2939GtoA | R980H	rs150748722	1	17313685	17313685
Parkinson disease, autosomal recessive	PINK1	92TtoA | L31X	N/A	1	20960133	20960133
Parkinson disease, autosomal recessive	PINK1	94GtoA | G32R	N/A	1	20960135	20960135
Parkinson disease, early-onset	PINK1	1391GtoA | R464H	N/A	1	20975627	20975627
Parkinson disease, early-onset	PINK1	1426GtoA | E476K	rs115477764	1	20975662	20975662
Parkinson disease, early-onset	PINK1	1557delG | 1557del1 | 1557del	N/A	1	20976995	20976995
Diabetes, type 2, increased risk	PINK1	1562AtoC | N521T	N/A	1	20977000	20977000
Parkinson disease, early-onset	PINK1	1573GtoA | D525N	N/A	1	20977011	20977011
Parkinson disease, early-onset	PINK1	1570_1573dupTTAG | 1570_1573dup4 | 1570_1573dup	N/A	1	20977011	20977012
Schwartz-Jampel syndrome type 1	HSPG2	4740GtoA	N/A	1	22190593	22190593
Schwartz-Jampel syndrome type 1	HSPG2	4648CtoT | R1550C	N/A	1	22190685	22190685
Primary amenorrhea with Mullerian Duct abn	WNT4	35TtoC | L12P	rs121908653	1	22469381	22469381
C1Q deficiency, type A	C1QA	2TtoG | M1R	N/A	1	22964111	22964111
C1Q deficiency, type A	C1QA	91GtoA | G31R	N/A	1	22964200	22964200
Diamond-Blackfan anaemia	RPL11	94_97delAGAC | 94_97del4 | 94_97del	N/A	1	24019186	24019189
Diamond-Blackfan anaemia	RPL11	95_96delGA | 95_96del2 | 95_96del	N/A	1	24019187	24019188
Diamond-Blackfan anaemia	RPL11	100dupA | 100dup1 | 100dup	N/A	1	24019192	24019193
Rhesus blood group variant	RHCE	149minus1GtoA	N/A	1	25735361	25735361
Hypercholesterolaemia, autosomal recessive	LDLRAP1	167CtoT | T56M	N/A	1	25880491	25880491
SEPN-related myopathy	SEPN1	166CtoT | Q56X	N/A	1	26126887	26126887
SEPN-related myopathy	SEPN1	183plus1GtoC	N/A	1	26126905	26126905
Lumbar stenosis	COL9A2	1369minus2AtoC	N/A	1	40769513	40769513
Glucose transporter 1 deficiency syndrome	SLC2A1	1100dupT | 1100dup1 | 1100dup	N/A	1	43393453	43393454
Glucose transporter 1 deficiency syndrome	SLC2A1	1080delG | 1080del1 | 1080del	N/A	1	43393474	43393474
Glucose transporter 1 deficiency syndrome	SLC2A1	506TtoC | L169P	N/A	1	43396307	43396307
Glucose transporter 1 deficiency syndrome	SLC2A1	497_499delTCG | 497_499del3 | 497_499del	N/A	1	43396314	43396316
Paroxysmal dyskinesia, exertion-induced, epile	SLC2A1	493GtoA | V165I	N/A	1	43396320	43396320
Amegakaryocytic thrombocytopaenia, congen	MPL	769CtoT | R257C	rs121913611	1	43805713	43805713
Amegakaryocytic thrombocytopaenia, congen	MPL	770GtoT | R257L	N/A	1	43805714	43805714
Macrostomia	PTCH2	1411GtoA | V471I	N/A	1	45294707	45294707
Nevoid basal cell carcinoma syndrome	PTCH2	1307CtoT | A436V	N/A	1	45294893	45294893
Porphyria, cutanea tarda	UROD	74GtoA | G25E	N/A	1	45478632	45478632
Porphyria, cutanea tarda	UROD	96delC | 96del1 | 96del	N/A	1	45478654	45478654
Porphyria, cutanea tarda	UROD	100TtoA | W34R	N/A	1	45478658	45478658
Porphyria, cutanea tarda	UROD	102GtoA | W34X	N/A	1	45478660	45478660
Porphyria, cutanea tarda	UROD	113AtoG | Q38R	N/A	1	45478671	45478671
Porphyria, cutanea tarda	UROD	121_122insCGGCC | 121_122ins5 | 121_122ins	N/A	1	45478679	45478680
Porphyria, cutanea tarda	UROD	126CtoG | Y42X	N/A	1	45478684	45478684
Porphyria, cutanea tarda	UROD	346CtoT | Q116X	N/A	1	45479335	45479335
Porphyria, cutanea tarda	UROD	358CtoT | R120C	N/A	1	45479347	45479347
Muscle-eye-brain disease	POMGNT1	1895plus1GtoA	N/A	1	46655129	46655129
Muscle-eye-brain disease	POMGNT1	1895plus1GtoT	N/A	1	46655129	46655129
Muscle-eye-brain disease	POMGNT1	1876delG | 1876del1 | 1876del	N/A	1	46655149	46655149
Muscle-eye-brain disease	POMGNT1	1864delC | 1864del1 | 1864del	N/A	1	46655161	46655161
Eye developmental anomalies, autosomal rece	FOXE3	21_24delGGAT | 21_24del4 | 21_24del	N/A	1	47882008	47882011
Eye developmental anomalies, autosomal don	FOXE3	146GtoC | G49A	N/A	1	47882133	47882133
Microphthalmia, nonsyndromic bilateral	FOXE3	705delC | 705del1 | 705del	N/A	1	47882692	47882692
Aphakia, congenital, primary	FOXE3	720CtoA | C240X	rs80358194	1	47882707	47882707
Carnitine palmitoyltransferase 2 deficiency	CPT2	38delG | 38del1 | 38del	N/A	1	53662653	53662653
Carnitine palmitoyltransferase 2 deficiency	CPT2	36_43dupGGGCCCCG | 36_43dup8 | 36_43dup	N/A	1	53662658	53662659
Carnitine palmitoyltransferase 2 deficiency	CPT2	98delA | 98del1 | 98del	N/A	1	53662713	53662713
Coronary heart disease, reduced risk, associat	PCSK9	137GtoT | R46L	rs11591147	1	55505647	55505647
Hypercholesterolaemia	PCSK9	158CtoT | A53V	rs11583680	1	55505668	55505668
High LDL cholesterol	PCSK9	161AtoC | E54A	N/A	1	55505671	55505671
Low LDL cholesterol, association with	PCSK9	169GtoA | E57K	rs145886902	1	55505679	55505679
Low LDL cholesterol	PCSK9	202delG | 202del1 | 202del	N/A	1	55505712	55505712
Leber congenital amaurosis	RPE65	90_91insT | 90_91ins1 | 90_91ins	N/A	1	68914310	68914311
Retinal dystrophy, severe, childhood onset	RPE65	89dupT | 89dup1 | 89dup	N/A	1	68914311	68914312
Retinal dystrophy, childhood onset	RPE65	74CtoT | P25L	N/A	1	68914327	68914327
Retinal dystrophy, autosomal recessive	RPE65	65TtoC | L22P	rs61751277	1	68914336	68914336
Leber congenital amaurosis	RPE65	57_58delGG | 57_58del2 | 57_58del	rs62642582	1	68914343	68914344
Leber congenital amaurosis	RPE65	16GtoT | E6X	N/A	1	68914385	68914385
Stargardt disease	ABCA4	4469GtoA | C1490Y	rs61751402	1	94495071	94495071
Stargardt disease	ABCA4	4467GtoT | R1489S	rs140952412	1	94495073	94495073
Stargardt disease	ABCA4	4463GtoA | C1488Y	N/A	1	94495077	94495077
Stargardt disease	ABCA4	4463GtoT | C1488F	rs61750147	1	94495077	94495077
Stargardt disease	ABCA4	4462TtoC | C1488R	rs61750146	1	94495078	94495078
Stargardt disease	ABCA4	4457CtoT | P1486L	rs61750145	1	94495083	94495083
Stargardt disease	ABCA4	4450CtoT | P1484S	N/A	1	94495090	94495090
Stargardt disease	ABCA4	4436GtoA | W1479X	rs61752434	1	94495104	94495104
Stargardt disease	ABCA4	4417CtoA | L1473M	rs142732109	1	94495123	94495123
Macular dystrophy, early-onset	ABCA4	4363TtoC | C1455R	N/A	1	94495177	94495177
Stargardt disease	ABCA4	4353minus1GtoT	rs61750144	1	94495188	94495188
Stargardt disease	ABCA4	4217AtoG | H1406R	N/A	1	94496588	94496588
Stargardt disease	ABCA4	4216CtoG | H1406D	N/A	1	94496589	94496589
Stargardt disease	ABCA4	4216CtoT | H1406Y	rs61750133	1	94496589	94496589

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Stargardt disease	ABCA4	4200CtoA | Y1400X	N/A	1	94496605	94496605
Stargardt disease	ABCA4	4195GtoA | E1399K	rs62642573	1	94496610	94496610
Stargardt disease	ABCA4	4177GtoA | V1393I	N/A	1	94496628	94496628
Cone-rod dystrophy	ABCA4	4169TtoC | L1390P	rs61752430	1	94496636	94496636
Stargardt disease	ABCA4	4163TtoC | L1388P	rs61750131	1	94496642	94496642
Stargardt disease	ABCA4	4139CtoT | P1380L	rs61750130	1	94496666	94496666
Pseudohermaphroditism	HSD3B2	299AtoG | N100S	N/A	1	119962197	119962197
Mitochondrial HMG-CoA synthase deficiency	HMGCS2	1499GtoA | R500H	rs137852639	1	120293453	120293453
Dyschromatosis symmetrica hereditaria	ADAR	3273delT | 3273del1 | 3273del	N/A	1	154558271	154558271
Dyschromatosis symmetrica hereditaria	ADAR	3272TtoC | F1091S	N/A	1	154558272	154558272
Dyschromatosis symmetrica hereditaria	ADAR	3248GtoA | R1083H	N/A	1	154558296	154558296
Dyschromatosis symmetrica hereditaria	ADAR	3247CtoT | R1083C	N/A	1	154558297	154558297
Dyschromatosis symmetrica hereditaria	ADAR	3241TtoA | C1081S	N/A	1	154558303	154558303
Dyschromatosis symmetrica hereditaria	ADAR	3232CtoT | R1078C	N/A	1	154558312	154558312
Dyschromatosis symmetrica hereditaria	ADAR	3222_3226delGCATC | 3222_3226del5 | 3222_3226del	N/A	1	154558318	154558322
Dyschromatosis symmetrica hereditaria	ADAR	3224AtoG | H1075R	N/A	1	154558320	154558320
Dyschromatosis symmetrica hereditaria	ADAR	3208dupC | 3208dup1 | 3208dup	N/A	1	154558335	154558336
Dyschromatosis symmetrica hereditaria	ADAR	3203GtoT | G1068V	N/A	1	154558341	154558341
Dyschromatosis symmetrica hereditaria	ADAR	3203minus2AtoG	N/A	1	154558343	154558343
Dyschromatosis symmetrica hereditaria	ADAR	2969_2970delCT | 2969_2970del2 | 2969_2970del	N/A	1	154560650	154560651
Dyschromatosis symmetrica hereditaria	ADAR	2969CtoG | P990R	N/A	1	154560651	154560651
Dyschromatosis symmetrica hereditaria	ADAR	2967CtoA | Y989X	N/A	1	154560653	154560653
Dyschromatosis symmetrica hereditaria	ADAR	2929delA | 2929del1 | 2929del	N/A	1	154560691	154560691
Pyruvate kinase deficiency	PKLR	173_188dupAGCAGCAGCTGCCAGC | 173_188dup16 | 173_188dup	N/A	1	155269983	155269984
Progeroid syndrome, atypical	LMNA	406GtoC | D136H	N/A	1	156100457	156100457
Muscular dystrophy, Emery-Dreifuss	LMNA	419TtoC | L140P	N/A	1	156100470	156100470
Werner syndrome, atypical	LMNA	419TtoG | L140R	rs60652225	1	156100470	156100470
Cardiomyopathy, dilated	LMNA	427TtoC | S143P	rs61661343	1	156100478	156100478
Myopathy, early-onset and progeria	LMNA	428CtoT | S143F	rs58912633	1	156100479	156100479
Hutchinson-Gilford progeria syndrome	LMNA	433GtoA | E145K	rs60310264	1	156100484	156100484
Muscular dystrophy, Emery-Dreifuss	LMNA	447dupC | 447dup1 | 447dup	N/A	1	156100498	156100499
Muscular dystrophy, Emery-Dreifuss	LMNA	448AtoC | T150P	rs58917027	1	156100499	156100499
Muscular dystrophy, Emery-Dreifuss	LMNA	800AtoG | Y267C	rs57048196	1	156104756	156104756
Muscular dystrophy, Emery-Dreifuss	LMNA	802TtoC | S268P	N/A	1	156104758	156104758
Muscular dystrophy, Emery-Dreifuss	LMNA	881AtoC | Q294P	rs61616775	1	156105048	156105048
Muscular dystrophy, Emery-Dreifuss	LMNA	883TtoC | S295P	N/A	1	156105050	156105050
Charcot-Marie-Tooth disease 2	LMNA	892CtoT | R298C	rs59885338	1	156105059	156105059
Partial lipodystrophy, Dunnigan	LMNA	895AtoG | I299V	rs150924946	1	156105062	156105062
Werner syndrome with ischemic disease	LMNA	898GtoA | D300N	N/A	1	156105065	156105065
Muscular dystrophy	LMNA	905TtoC | L302P	N/A	1	156105072	156105072
Pain insensitivity, congenital	NTRK1	924_930delGCCGGCA | 924_930del7 | 924_930del	N/A	1	156843498	156843504
Poikilocytic anaemia	SPTA1	3188plus5GtoA	N/A	1	158623059	158623059
Type 2 diabetes, association with	KCNJ9	1097TtoC | V366A	rs3001040	1	160057522	160057522
Neural tube defects	VANGL2	1310TtoC | F437S	N/A	1	160394912	160394912
Polyneuropathy	MPZ	106AtoT | R36W	N/A	1	161277176	161277176
Charcot-Marie-Tooth disease	MPZ	101CtoT | T34I	N/A	1	161277181	161277181
Charcot-Marie-Tooth disease	MPZ	98AtoT | Y33F	N/A	1	161277184	161277184
Charcot-Marie-Tooth disease 1b	MPZ	94GtoT | V32F	N/A	1	161277188	161277188
Charcot-Marie-Tooth disease 1b	MPZ	90CtoG | I30M	N/A	1	161277192	161277192
Dejerine-Sottas syndrome	MPZ	89TtoC | I30T	N/A	1	161277193	161277193
Charcot-Marie-Tooth disease	MPZ	89TtoG | I30S	N/A	1	161277193	161277193
Charcot-Marie-Tooth disease 1b	MPZ	88_89delATinsGGGGTTTACAC | 88_89del2ins11 | 88_89delins	N/A	1	161277193	161277194
Charcot-Marie-Tooth disease 2	MPZ	59CtoT | S20F	N/A	1	161279637	161279637
ACTH deficiency, isolated	TBX19	477GtoC | L159F	N/A	1	168262390	168262390
ACTH deficiency, isolated	TBX19	512TtoC | I171T	N/A	1	168262425	168262425
ACTH deficiency, isolated	TBX19	535CtoT | R179X	N/A	1	168262448	168262448
Megaloblastic anaemia, thiamine responsive	SLC19A2	759dupT | 759dup1 | 759dup	N/A	1	169446440	169446441
Megaloblastic anaemia, thiamine responsive	SLC19A2	759delT | 759del1 | 759del	N/A	1	169446441	169446441
Megaloblastic anaemia, thiamine responsive	SLC19A2	750GtoA | W250X	rs74315374	1	169446450	169446450
Factor V deficiency	F5	6293CtoT | P2098L	N/A	1	169487702	169487702
Primary microcephaly	ASPM	9238AtoT | K3080X	N/A	1	197062238	197062238
Ectodermal dysplasia/skin fragility syndrome	PKP1	889delC | 889del1 | 889del	N/A	1	201286742	201286742
Ectodermal dysplasia/skin fragility syndrome	PKP1	897_901delCAACC | 897_901del5 | 897_901del	N/A	1	201286750	201286754
Ectodermal dysplasia/skin fragility syndrome	PKP1	910CtoT | Q304X	rs121918354	1	201286763	201286763
Cardiomyopathy, hypertrophic	TNNT2	860GtoA | W287X	N/A	1	201328345	201328345
Cardiomyopathy, hypertrophic	TNNT2	780plus8GtoA	N/A	1	201330399	201330399
Cardiomyopathy, hypertrophic	TNNT2	571_573delCAG | 571_573del3 | 571_573del	N/A	1	201331520	201331522
Reduced enzyme activity	CHIT1	1060GtoA | G354R	rs9943208	1	203186963	203186963
Ulcerative colitis, association with	IL10	386TtoA | F129Y	N/A	1	206943232	206943232
Epidermolysis bullosa, Herlitz	LAMB3	1188_1198delCGGGCAGTGTG | 1188_1198del11 | 1188_1198del	N/A	1	209801470	209801480
Van der Woude syndrome	IRF6	69CtoA | Y23X	N/A	1	209974690	209974690
Van der Woude syndrome	IRF6	65TtoC | L22P	N/A	1	209974694	209974694
Van der Woude syndrome	IRF6	49_64delCAGGTGGATAGTGGCC | 49_64del16 | 49_64del	N/A	1	209974695	209974710
Hypospadias	ATF3	271AtoG | R91G	N/A	1	212791499	212791499
Usher syndrome 2	USH2A	6169CtoT | Q2057X	N/A	1	216219929	216219929
Usher syndrome 2a	USH2A	3840_3841delGAinsCT | 3840_3841del2ins2 | 3840_3841delins	N/A	1	216371897	216371898
Usher syndrome 2	USH2A	2996_3009delGTCAGCCTTGTAAT | 2996_3009del14 | 2996_3009del	N/A	1	216390877	216390890
Usher syndrome 2a	USH2A	3004delT | 3004del1 | 3004del	N/A	1	216390882	216390882
Pelizaeus-Merzbacher-like disease	GJC2	373CtoT | R125X	N/A	1	228345832	228345832
Lymphoedema	GJC2	374GtoA | R125Q	N/A	1	228345833	228345833
Pelizaeus-Merzbacher-like disease	GJC2	392delC | 392del1 | 392del	N/A	1	228345851	228345851
Pelizaeus-Merzbacher-like disease	GJC2	445GtoA | G149S	N/A	1	228345904	228345904
Nemaline myopathy	ACTA1	16GtoA | E6K	N/A	1	229568847	229568847
Cardiomyopathy, hypertrophic	ACTN2	1748AtoC | E583A	N/A	1	236914861	236914861
Hypothyroidism	TPO	1274AtoG | N425S	N/A	2	1481312	1481312
Rhabdomyolysis	LPIN1	508_509delGA | 508_509del2 | 508_509del	N/A	2	11911717	11911718
Obesity, adrenal insufficiency and red hair	POMC	206delC | 206del1 | 206del	N/A	2	25384548	25384548
Deafness, non-syndromic	OTOF	1740delC | 1740del1 | 1740del	N/A	2	26703717	26703717
Deafness, autosomal recessive 9	OTOF	1718TtoG | L573R	N/A	2	26703739	26703739
Leukoencephalopathy with vanishing white m	EIF2B4	1117CtoT | R373C	rs113994035	2	27589697	27589697
Leukoencephalopathy with vanishing white m	EIF2B4	1088GtoA | R363Q	rs113994034	2	27589726	27589726
Decreased activity	XDH	1663CtoT | P555S	rs45577338	2	31596762	31596762
Xanthinuria, type 1	XDH	445CtoT | R149C	rs72549369	2	31620584	31620584
Spastic paraplegia	SPAST	1099-2_1099delAGT | 1099-2_1099del3 | 1099-2_1099del	N/A	2	32352015	32352017
Spastic paraplegia	SPAST	1101_1103delGTT | 1101_1103del3 | 1101_1103del	N/A	2	32352019	32352021

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Spastic paraplegia, autosomal dominant	SPAST	1103TtoC | F368S	N/A	2	32352021	32352021
Spastic paraplegia	SPAST	1104CtoA | F368L	N/A	2	32352022	32352022
Spastic paraplegia, autosomal dominant	SPAST	1105dupA | 1105dup1 | 1105dup	N/A	2	32352023	32352024
Spastic paraplegia, autosomal dominant	SPAST	1108GtoA | G370R	N/A	2	32352026	32352026
Spastic paraplegia, autosomal dominant	SPAST	1114AtoT | R372X	N/A	2	32352032	32352032
Spastic paraplegia, autosomal dominant	SPAST	1115delG | 1115del1 | 1115del	N/A	2	32352033	32352033
Spastic paraplegia	SPAST	1115dupG | 1115dup1 | 1115dup	N/A	2	32352033	32352034
Spastic paraplegia	SPAST	1120CtoA | P374T	N/A	2	32352038	32352038
Spastic paraplegia	SPAST	1121CtoG | P374R	N/A	2	32352039	32352039
Spastic paraplegia	SPAST	1130GtoA | G377E	N/A	2	32352048	32352048
Spastic paraplegia, autosomal dominant	SPAST	1133TtoA | L378Q	N/A	2	32352051	32352051
Spastic paraplegia	SPAST	1133TtoG | L378R	N/A	2	32352051	32352051
Spastic paraplegia, autosomal dominant	SPAST	1134_1135delGT | 1134_1135del2 | 1134_1135del	N/A	2	32352052	32352053
Spastic paraplegia	SPAST	1136dupT | 1136dup1 | 1136dup	N/A	2	32352054	32352055
Spastic paraplegia, autosomal dominant	SPAST	1137_1142delACTCTT | 1137_1142del6 | 1137_1142del	N/A	2	32352055	32352060
Spastic paraplegia, autosomal dominant	SPAST	1137dupA | 1137dup1 | 1137dup	N/A	2	32352055	32352056
Spastic paraplegia, autosomal dominant	SPAST	1139TtoA | L380H	N/A	2	32352057	32352057
Spastic paraplegia, autosomal dominant	SPAST	1141TtoC | F381L	N/A	2	32352059	32352059
Spastic paraplegia, autosomal dominant	SPAST	1142TtoG | F381C	N/A	2	32352060	32352060
Spastic paraplegia	SPAST	1144GtoC | G382R	N/A	2	32352062	32352062
Spastic paraparesis	SPAST	1153GtoT | G385W	N/A	2	32352071	32352071
Spastic paraplegia	SPAST	1154GtoA | G385E	N/A	2	32352072	32352072
Spastic paraplegia, autosomal dominant	SPAST	1157AtoG | N386S	rs121908514	2	32352075	32352075
Spastic paraplegia, autosomal dominant	SPAST	1158TtoG | N386K	N/A	2	32352076	32352076
Spastic paraplegia, autosomal dominant	SPAST	1160_1170delGGAAGACAATG | 1160_1170del11 | 1160_1170del	N/A	2	32352078	32352088
Spastic paraplegia, autosomal dominant	SPAST	1163AtoG | K388R	N/A	2	32352081	32352081
Spastic paraplegia, autosomal dominant	SPAST	1167_1168delAA | 1167_1168del2 | 1167_1168del	N/A	2	32352085	32352086
Spastic paraplegia, autosomal dominant	SPAST	1168AtoG | M390V	N/A	2	32352086	32352086
Spastic paraparesis	SPAST	1172TtoC | L391P	N/A	2	32352090	32352090
Spastic paraplegia, autosomal dominant	SPAST	1173plus1GtoA	N/A	2	32352092	32352092
Glaucoma, primary congenital	CYP1B1	1136CtoT | P379L	rs56305281	2	38298361	38298361
Glaucoma, primary congenital	CYP1B1	1133TtoA | L378Q	N/A	2	38298364	38298364
Glaucoma, primary congenital	CYP1B1	1122CtoG | D374E	N/A	2	38298375	38298375
Glaucoma, primary congenital	CYP1B1	1120GtoA | D374N	rs104893622	2	38298377	38298377
Glaucoma, primary congenital	CYP1B1	1109delC | 1109del1 | 1109del	N/A	2	38298388	38298388
Glaucoma, primary congenital	CYP1B1	1103GtoA | R368H	rs79204362	2	38298394	38298394
Glaucoma, primary congenital	CYP1B1	1103GtoT | R368L	N/A	2	38298394	38298394
Glaucoma, primary congenital	CYP1B1	1102CtoT | R368C	rs72480442	2	38298395	38298395
Glaucoma, primary congenital	CYP1B1	1099delG | 1099del1 | 1099del	N/A	2	38298398	38298398
Glaucoma, primary congenital	CYP1B1	1093GtoT | G365W	rs55771538	2	38298404	38298404
Glaucoma, primary congenital	CYP1B1	1090GtoA | V364M	rs72549379	2	38298407	38298407
Glaucoma, primary congenital	CYP1B1	1088TtoA | V363D	N/A	2	38298409	38298409
Glaucoma, primary congenital	CYP1B1	454_455delAT | 454_455del2 | 454_455del	N/A	2	38302077	38302078
Sitosterolaemia	ABCG5	727CtoT | R243X	rs119479066	2	44053568	44053568
Sitosterolaemia	ABCG5	335dupA | 335dup1 | 335dup	N/A	2	44059152	44059153
Gastric cancer	MSH2	49GtoT | V17F	rs63750966	2	47630379	47630379
Colorectal cancer	MSH2	72_73insC | 72_73ins1 | 72_73ins	N/A	2	47630402	47630403
Colorectal cancer, non-polyposis	MSH2	82GtoT | E28X	rs63751246	2	47630412	47630412
Colorectal cancer, non-polyposis	MSH2	82delG | 82del1 | 82del	N/A	2	47630412	47630412
Colorectal cancer, non-polyposis	MSH2	94_103delACCACAGTGC | 94_103del10 | 94_103del	rs63750728	2	47630424	47630433
Colorectal cancer, non-polyposis	MSH2	97AtoC | T33P	rs63751107	2	47630427	47630427
Colorectal cancer, non-polyposis	MSH2	97AtoG | T33A	rs63751107	2	47630427	47630427
Colorectal cancer, non-polyposis	MSH2	110delT | 110del1 | 110del	rs63751056	2	47630440	47630440
Colorectal cancer	MSH2	118GtoA | G40S	rs63751260	2	47630448	47630448
Colorectal cancer, non-polyposis	MSH2	119delG | 119del1 | 119del	rs63750984	2	47630449	47630449
Colorectal cancer, non-polyposis	MSH2	128AtoG | Y43C	rs17217723	2	47630458	47630458
Colorectal cancer, non-polyposis	MSH2	131CtoT | T44M	N/A	2	47630461	47630461
Colorectal cancer, non-polyposis	MSH2	134CtoT | A45V	rs63750285	2	47630464	47630464
Colorectal cancer, non-polyposis	MSH2	135_136delGC | 135_136del2 | 135_136del	N/A	2	47630465	47630466
Colorectal cancer, non-polyposis	MSH2	137dupA | 137dup1 | 137dup	N/A	2	47630467	47630468
Colorectal cancer	MSH2	138CtoG | H46Q	rs33946261	2	47630468	47630468
Colorectal cancer, non-polyposis	MSH2	142GtoT | E48X	rs63750615	2	47630472	47630472
Muir-Torre syndrome	MSH2	145delG | 145del1 | 145del	rs63750644	2	47630475	47630475
Colorectal cancer, non-polyposis	MSH2	145_146delGA | 145_146del2 | 145_146del	rs63750334	2	47630475	47630476
Colorectal cancer, non-polyposis	MSH2	145_148delGACG | 145_148del4 | 145_148del	N/A	2	47630475	47630478
Colorectal cancer, non-polyposis	MSH2	1243_1246delCCTA | 1243_1246del4 | 1243_1246del	N/A	2	47657047	47657050
Colorectal cancer, non-polyposis	MSH2	1249delG | 1249del1 | 1249del	N/A	2	47657053	47657053
Colorectal cancer	MSH2	1255CtoA | Q419K	rs186103499	2	47657059	47657059
Colorectal cancer, non-polyposis	MSH2	1255CtoT | Q419X	N/A	2	47657059	47657059
Colorectal cancer, non-polyposis	MSH2	1683delA | 1683del1 | 1683del	N/A	2	47698125	47698125
Colorectal cancer, non-polyposis	MSH2	1685AtoT | E562V	N/A	2	47698127	47698127
Colorectal cancer, non-polyposis	MSH2	1687dupT | 1687dup1 | 1687dup	N/A	2	47698129	47698130
Colorectal cancer, non-polyposis	MSH2	1688AtoC | Y563S	N/A	2	47698130	47698130
Colorectal cancer, non-polyposis	MSH2	1690AtoG | T564A	rs55778204	2	47698132	47698132
Colorectal cancer, non-polyposis	MSH2	1696_1697delAA | 1696_1697del2 | 1696_1697del	N/A	2	47698138	47698139
Colorectal cancer, non-polyposis	MSH2	1696_1697delAAinsG | 1696_1697del2ins1 | 1696_1697delins	N/A	2	47698138	47698139
Colorectal cancer, non-polyposis	MSH2	1699AtoT | K567X	N/A	2	47698141	47698141
Muir-Torre syndrome	MSH2	1700_1704delAAACA | 1700_1704del5 | 1700_1704del	N/A	2	47698142	47698146
Colorectal cancer, non-polyposis	MSH2	1702dupA | 1702dup1 | 1702dup	N/A	2	47698144	47698145
Colorectal cancer, non-polyposis	MSH2	1705_1706delGA | 1705_1706del2 | 1705_1706del	rs63750393	2	47698147	47698148
Colorectal cancer, non-polyposis	MSH2	1705_1706dupGA | 1705_1706dup2 | 1705_1706dup	N/A	2	47698148	47698149
Colorectal cancer, non-polyposis	MSH2	1710TtoG | Y570X	N/A	2	47698152	47698152
Colorectal cancer, non-polyposis	MSH2	1717delG | 1717del1 | 1717del	N/A	2	47698159	47698159
Colorectal cancer, non-polyposis	MSH2	1720CtoT | Q574X	rs63751298	2	47698162	47698162
Colorectal cancer, non-polyposis	MSH2	1720delC | 1720del1 | 1720del	N/A	2	47698162	47698162
Colorectal cancer, non-polyposis	MSH2	1723delG | 1723del1 | 1723del	N/A	2	47698165	47698165
Colorectal cancer, early onset	MSH2	1730TtoC | I577T	rs63749910	2	47698172	47698172
Breast carcinoma	MSH2	1737dupA | 1737dup1 | 1737dup	N/A	2	47698179	47698180
Glioma	MSH2	1738GtoT | E580X	N/A	2	47698180	47698180
Colorectal cancer, non-polyposis	MSH2	1748AtoG | N583S	N/A	2	47698190	47698190
Colorectal cancer, non-polyposis	MSH2	1748AtoT | N583I	N/A	2	47698190	47698190
Colorectal cancer, non-polyposis	MSH2	1759GtoC | G587R	N/A	2	47698201	47698201
Colorectal cancer, non-polyposis	MSH2	1759GtoC	N/A	2	47698201	47698201
Colorectal cancer, non-polyposis	MSH2	2080_2081insT | 2080_2081ins1 | 2080_2081ins	N/A	2	47698201	47698202
Colorectal cancer, non-polyposis	MSH2	1759plus2TtoA	N/A	2	47698203	47698203

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Colorectal cancer, non-polyposis	MSH2	1759plus2TtoC	N/A	2	47698203	47698203
Colorectal cancer	MSH6	814GtoT | E272X	rs63750552	2	48025936	48025936
Multiple colorectal adenoma	MSH6	866_867delGCinsAA | 866_867del2ins2 | 866_867delins	N/A	2	48025988	48025989
Breast cancer, earlier onset, association with	LHCGR	50_55dupTGCAGC | 50_55dup6 | 50_55dup	rs4539842	2	48982755	48982756
Amenorrhoea and recurrent pregnancy loss	LHCGR	55_56insTGCCGC | 55_56ins6 | 55_56ins	N/A	2	48982755	48982756
Zellweger syndrome H	PEX13	702GtoA | W234X	rs104893661	2	61259163	61259163
Restless legs syndrome	MEIS1	815GtoA | R272H	rs61752693	2	66739353	66739353
Congenital myasthenic syndrome, limb-girdle	GFPT1	362TtoC | I121T	N/A	2	69586446	69586446
Congenital myasthenic syndrome, limb-girdle	GFPT1	128AtoT | D43V	N/A	2	69597228	69597228
Muscular dystrophy, limb girdle	DYSF	463GtoA | G155R	N/A	2	71740851	71740851
Muscular dystrophy, limb girdle / Miyoshi myo	DYSF	755CtoT | T252M	N/A	2	71742844	71742844
Dysferlinopathy	DYSF	757CtoT | R253W	rs149827237	2	71742846	71742846
Miyoshi myopathy	DYSF	1129CtoT | R377X	N/A	2	71753425	71753425
Miyoshi myopathy	DYSF	1758CtoA | Y586X	N/A	2	71780146	71780146
Miyoshi myopathy	DYSF	1795_1798dupTACT | 1795_1798dup4 | 1795_1798dup	N/A	2	71780186	71780187
Miyoshi myopathy	DYSF	1813CtoT | Q605X	rs121908953	2	71780201	71780201
Muscular dystrophy, limb girdle 2B	DYSF	3443minus33AtoG	N/A	2	71817308	71817308
Muscular dystrophy, limb girdle	DYSF	4872delG | 4872del1 | 4872del	N/A	2	71887767	71887767
Muscular dystrophy	DYSF	4872_4876delGCCCGinsCCCC | 4872_4876del5ins4 | 4872_4876delins	N/A	2	71887767	71887771
Dysferlinopathy	DYSF	4876delG | 4876del1 | 4876del	N/A	2	71887771	71887771
Muscular dystrophy, limb girdle 2B	DYSF	4882GtoA | G1628R	N/A	2	71887777	71887777
Muscular dystrophy, limb girdle 2B	DYSF	4883GtoT | G1628V	N/A	2	71887778	71887778
Dysferlinopathy	DYSF	5289GtoC | E1763D	N/A	2	71894594	71894594
Miyoshi myopathy	DYSF	5824TtoG | C1942G	N/A	2	71906243	71906243
Dysferlinopathy	DYSF	5836_5839delCAGC | 5836_5839del4 | 5836_5839del	N/A	2	71906255	71906258
HyperCKemia	DYSF	5871_5872delGT | 5871_5872del2 | 5871_5872del	N/A	2	71906290	71906291
Dysferlinopathy	DYSF	5884CtoT | Q1962X	N/A	2	71906303	71906303
Dysferlinopathy	DYSF	5903GtoA | W1968X	N/A	2	71906322	71906322
Muscular dystrophy, limb girdle	DYSF	5908CtoT | P1970S	N/A	2	71906327	71906327
Miyoshi myopathy	DYSF	5939TtoA | I1980K	N/A	2	71906358	71906358
Miyoshi myopathy	DYSF	5946plus1GtoA	N/A	2	71906366	71906366
Tetrahydrobiopterin deficiency	SPR	304GtoT	N/A	2	73114865	73114865
Vitamin-K-dependent coagulation factor defici	GGCX	2110CtoT | R704X	N/A	2	85777224	85777224
Surfactant protein B deficiency	SFTPB	790CtoT | R264C	N/A	2	85890889	85890889
Surfactant protein B deficiency	SFTPB	780CtoA | C260X	N/A	2	85890899	85890899
Surfactant protein B deficiency	SFTPB	74TtoC | L25P	N/A	2	85895269	85895269
Increased promoter activity	SFTPB	NULL	rs2077079	2	85895338	85895338
Spastic paraplegia 31	REEP1	105plus6TtoC	N/A	2	86509287	86509287
Phaeochromocytoma	TMEM127	409plus1GtoT	N/A	2	96920570	96920570
Phaeochromocytoma	TMEM127	117_120delGTCT | 117_120del4 | 117_120del	N/A	2	96931000	96931003
Phaeochromocytoma	TMEM127	113CtoA | A38D	N/A	2	96931007	96931007
Phaeochromocytoma	TMEM127	76CtoT | Q26X	N/A	2	96931044	96931044
Lower testosterone levels	NPAS2	1180GtoA | A394T	rs2305160	2	101591304	101591304
Ectodermal dysplasia, hypohidrotic	EDAR	719_722delAAGA | 719_722del4 | 719_722del	N/A	2	109527240	109527243
Leber congenital amaurosis	MERTK	2237AtoG | K746R	N/A	2	112779046	112779046
Retinitis pigmentosa	MERTK	2323CtoT | R775X	N/A	2	112779132	112779132
Craniofacial anomalies	GLI2	1885GtoA | E629K	N/A	2	121742248	121742248
Holoprosencephaly with anophthalmia, branc	GLI2	2794CtoT | P932S	N/A	2	121746284	121746284
Protein C deficiency	PROC	124CtoA | R42S	N/A	2	128178912	128178912
Protein C deficiency	PROC	124CtoT | R42C	N/A	2	128178912	128178912
Protein C deficiency	PROC	125GtoA | R42H	N/A	2	128178913	128178913
Protein C deficiency	PROC	125GtoT | R42L	N/A	2	128178913	128178913
Protein C deficiency	PROC	127GtoA | A43T	N/A	2	128178915	128178915
Protein C deficiency	PROC	131AtoT | N44I	N/A	2	128178919	128178919
Protein C deficiency	PROC	143delA | 143del1 | 143del	N/A	2	128178931	128178931
Protein C deficiency	PROC	147GtoT | E49D	N/A	2	128178935	128178935
Protein C deficiency	PROC	151CtoT | R51C	N/A	2	128178939	128178939
Protein C deficiency	PROC	152GtoA | R51H	N/A	2	128178940	128178940
Protein C deficiency, type II	PROC	157AtoC | S53R	N/A	2	128178945	128178945
Protein C deficiency	PROC	160AtoT | S54C	N/A	2	128178948	128178948
Protein C deficiency, type II	PROC	162CtoG | S54R	N/A	2	128178950	128178950
Protein C deficiency	PROC	164TtoC | L55P	N/A	2	128178952	128178952
Protein C deficiency	PROC	169CtoG | R57G	N/A	2	128178957	128178957
Protein C deficiency	PROC	169CtoT | R57W	N/A	2	128178957	128178957
Protein C deficiency	PROC	170GtoA | R57Q	N/A	2	128178958	128178958
Protein C deficiency	PROC	177CtoA | C59X	N/A	2	128178965	128178965
Protein C deficiency	PROC	185AtoC | E62A	rs121918148	2	128178973	128178973
Protein C deficiency	PROC	199GtoA | E67K	N/A	2	128178987	128178987
Protein C deficiency	PROC	199GtoC | E67Q	N/A	2	128178987	128178987
Protein C deficiency	PROC	202GtoA | E68K	N/A	2	128178990	128178990
Protein C deficiency	PROC	208AtoG | K70E	N/A	2	128178996	128178996
Protein C deficiency, type II	PROC	211GtoA | E71K	N/A	2	128178999	128178999
Protein C deficiency	PROC	211GtoT | E71X	N/A	2	128178999	128178999
Protein C deficiency	PROC	226GtoA | V76M	rs121918149	2	128179014	128179014
Protein C deficiency	PROC	230AtoG | D77G	N/A	2	128179018	128179018
Protein C deficiency	PROC	237plus3AtoG	N/A	2	128179028	128179028
Protein C deficiency	PROC	237plus5GtoA	N/A	2	128179030	128179030
Protein C deficiency	PROC	237plus15GtoA	N/A	2	128179040	128179040
Protein C deficiency	PROC	401-18_401-3delGCCCTCCCCTGCCCGC | 401-18_401-3del16 | 401-18_401-3del	N/A	2	128180832	128180847
Protein C deficiency	PROC	401minus2AtoG	N/A	2	128180848	128180848
Protein C deficiency	PROC	418TtoC | C140R	N/A	2	128180867	128180867
Protein C deficiency	PROC	422CtoA | S141X	N/A	2	128180871	128180871
Protein C deficiency	PROC	440GtoA | C147Y	N/A	2	128180889	128180889
Protein C deficiency	PROC	441CtoA | C147X	N/A	2	128180890	128180890
Protein C deficiency	PROC	785TtoC | L262P	N/A	2	128184787	128184787
Protein C deficiency	PROC	786dupT | 786dup1 | 786dup	N/A	2	128184788	128184789
Protein C deficiency	PROC	793CtoT | L265F	rs121918156	2	128184795	128184795
Protein C deficiency	PROC	796plus1GtoT	N/A	2	128184799	128184799
Homocystinuria, cblD type	MMADHC	683CtoG | S228X	N/A	2	150427612	150427612
Nemaline myopathy	NEB	11042_11059delTCCTAGCAAAGAATAATG | 11042_11059del18 | 11042_11059del	N/A	2	152468717	152468734
Nemaline myopathy	NEB	78plus1GtoA	N/A	2	152586128	152586128
Dravet syndrome C	SCN1A	698TtoG | L233R	N/A	2	166908495	166908495
Dravet syndrome	SCN1A	697_698delCT | 697_698del2 | 697_698del	N/A	2	166908495	166908496
Intrahepatic cholestasis, familial progressive 2	ABCB11	2576CtoG | T859R	N/A	2	169801149	169801149
Cholestasis, drug-induced	ABCB11	2563GtoA | G855R	N/A	2	169801162	169801162

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Donnai-Barrow syndrome	LRP2	11469_11472delTTTG | 11469_11472del4 | 11469_11472del	rs80338753	2	170026237	170026240
Altered enzyme activity	PDE11A	2458GtoA | V820M	rs140269105	2	178540212	178540212
Ehlers-Danlos syndrome, vascular-type	COL3A1	709GtoA | G237R	N/A	2	189854840	189854840
Ehlers-Danlos syndrome IV	COL3A1	718GtoC | G240R	N/A	2	189854849	189854849
Ehlers-Danlos syndrome IV	COL3A1	728GtoT | G243V	N/A	2	189854859	189854859
Ehlers-Danlos syndrome, vascular-type	COL3A1	1511GtoA | G504D	N/A	2	189860419	189860419
Ehlers-Danlos syndrome, vascular-type	COL3A1	1550CtoT | P517L	rs142085247	2	189860458	189860458
Ehlers-Danlos syndrome	COL3A1	1925GtoA | G642D	N/A	2	189862993	189862993
Ehlers-Danlos syndrome IV	COL3A1	3202minus2AtoG	N/A	2	189871661	189871661
Ehlers-Danlos syndrome IV	COL3A1	3212GtoT | G1071V	N/A	2	189871673	189871673
Ehlers-Danlos syndrome IV	COL3A1	3230GtoT | G1077V	rs121912915	2	189871691	189871691
Ehlers-Danlos syndrome IV	COL3A1	3221_3235dupGTGCTCCCGGTCCTG | 3221_3235dup15 | 3221_3235dup	N/A	2	189871696	189871697
Ehlers-Danlos syndrome IV	COL3A1	3255plus1GtoA	N/A	2	189871717	189871717
Ehlers-Danlos syndrome IV	COL3A1	3255plus5GtoA	N/A	2	189871721	189871721
Neurodegeneration, progressive infantile	HIBCH	365AtoG | Y122C	rs121918329	2	191155151	191155151
Carbamoyl phosphate synthetase I deficiency	CPS1	236GtoA | G79E	N/A	2	211438131	211438131
Carbamoyl phosphate synthetase I deficiency	CPS1	236plus6TtoC	N/A	2	211438137	211438137
Carbamoyl phosphate synthetase I deficiency	CPS1	2719_2721delGCAinsCC | 2719_2721del3ins2 | 2719_2721delins	N/A	2	211502457	211502459
Carbamoyl phosphate synthetase I deficiency	CPS1	2795GtoC | R932T	N/A	2	211502533	211502533
Carbamoyl phosphate synthetase I deficiency	CPS1	2798delT | 2798del1 | 2798del	N/A	2	211502536	211502536
Carbamoyl phosphate synthetase I deficiency	CPS1	2809_2810delAT | 2809_2810del2 | 2809_2810del	N/A	2	211502547	211502548
Carbamoyl phosphate synthetase I deficiency	CPS1	2993CtoT | S998F	N/A	2	211507241	211507241
Carbamoyl phosphate synthetase I deficiency	CPS1	2996_2998delGTA | 2996_2998del3 | 2996_2998del	N/A	2	211507244	211507246
Carbamoyl phosphate synthetase I deficiency	CPS1	3037_3039delGTG | 3037_3039del3 | 3037_3039del	N/A	2	211507285	211507287
Carbamoyl phosphate synthetase I deficiency	CPS1	3044dupG | 3044dup1 | 3044dup	N/A	2	211507292	211507293
Carbamoyl phosphate synthetase I deficiency	CPS1	3047AtoG | N1016S	N/A	2	211507295	211507295
Carbamoyl phosphate synthetase I deficiency	CPS1	3050CtoT | P1017L	N/A	2	211507298	211507298
3-M syndrome	OBSL1	1119GtoC | W373C	N/A	2	220432940	220432940
3-M syndrome	OBSL1	1039dupC | 1039dup1 | 1039dup	N/A	2	220433019	220433020
Waardenburg syndrome	PAX3	284_289dupCCATAC | 284_289dup6 | 284_289dup	N/A	2	223161728	223161729
Waardenburg syndrome	PAX3	288delA | 288del1 | 288del	N/A	2	223161730	223161730
Waardenburg syndrome	PAX3	268_281delTACCAGGAGACTGG | 268_281del14 | 268_281del	N/A	2	223161737	223161750
Waardenburg syndrome III	PAX3	268TtoC | Y90H	rs104893654	2	223161750	223161750
Microhaematuria and proteinuria	COL4A3	1AtoT | M1L	N/A	2	228029443	228029443
Alport syndrome	COL4A3	3752minus2AtoT	N/A	2	228163396	228163396
Alport syndrome	COL4A3	3829GtoA | G1277S	rs190598500	2	228163475	228163475
Alport syndrome	COL4A3	3882plus5GtoC	N/A	2	228163533	228163533
Alport syndrome	COL4A3	4231AtoT | K1411X	N/A	2	228169778	228169778
Pterygium syndrome	CHRNG	13CtoT | Q5X	N/A	2	233404470	233404470
Pterygium syndrome	CHRNG	55GtoA | G19R	N/A	2	233404512	233404512
Crohn's disease, increased risk, association wit	ATG16L1	841AtoG | T281A	rs2241880	2	234183368	234183368
Crigler-Najjar syndrome 2	UGT1A1	44TtoG | L15R	rs111033541	2	234668977	234668977
Bethlem myopathy	COL6A3	6166GtoA | G2056R	N/A	2	238269808	238269808
Collagen VI deficiency	COL6A3	6157minus2AtoG	N/A	2	238269819	238269819
Griscelli syndrome	MLPH	103CtoT | R35W	rs119473031	2	238402172	238402172
Hyperoxaluria	AGXT	364CtoT | R122X	rs180177210	2	241810066	241810066
Hyperoxaluria	AGXT	385GtoC | D129H	rs180177212	2	241810087	241810087
Hyperoxaluria	AGXT	409CtoT | Q137X	rs180177214	2	241810111	241810111
Hyperoxaluria	AGXT	416_418delTGG | 416_418del3 | 416_418del	rs180177215	2	241810118	241810120
Hyperoxaluria	AGXT	423GtoT | E141D	rs180177217	2	241810125	241810125
Hyperoxaluria	AGXT	1125_1126delCG | 1125_1126del2 | 1125_1126del	rs180177164	2	241818184	241818185
Hyperoxaluria	AGXT	1151TtoC | L384P	rs180177165	2	241818210	241818210
Potential protein deficiency	PASK	3799CtoT | R1267X	N/A	2	242046783	242046783
D-2-hydroxyglutaric aciduria	D2HGDH	1331TtoC | V444A	rs121434360	2	242707149	242707149
D-2-hydroxyglutaric aciduria	D2HGDH	1333_1334delAC | 1333_1334del2 | 1333_1334del	N/A	2	242707151	242707152
D-2-hydroxyglutaric aciduria	D2HGDH	1337CtoT | A446V	N/A	2	242707155	242707155
Multiple sulphatase deficiency	SUMF1	243delC | 243del1 | 243del	N/A	3	4508687	4508687
Cardiac atrioventricular septal defect	CRELD1	320GtoA | R107H	rs28941780	3	9979308	9979308
Cardiac atrioventricular septal defect	CRELD1	932CtoT | T311I	rs28942092	3	9985083	9985083
Bladder cancer, increased risk	XPC	905TtoC | F302S	rs121965091	3	14201326	14201326
Endplate acetylcholinesterase deficiency	COLQ	1298plus3AtoG	N/A	3	15495333	15495333
Endplate acetylcholinesterase deficiency	COLQ	506CtoG | S169X	rs104893734	3	15516954	15516954
Endplate acetylcholinesterase deficiency	COLQ	219plus1GtoC	rs149020371	3	15531031	15531031
Endplate acetylcholinesterase deficiency	COLQ	176CtoA | P59Q	N/A	3	15531075	15531075
Endplate acetylcholinesterase deficiency	COLQ	175CtoT | P59S	N/A	3	15531076	15531076
Endplate acetylcholinesterase deficiency	COLQ	157dupC | 157dup1 | 157dup	N/A	3	15531093	15531094
Gangliosidosis GM1	GLB1	452AtoT | D151V	N/A	3	33109727	33109727
Gangliosidosis GM1	GLB1	451GtoT | D151Y	N/A	3	33109728	33109728
Morquio disease B	GLB1	446CtoT | S149F	N/A	3	33109733	33109733
Gangliosidosis GM1	GLB1	188dupT | 188dup1 | 188dup	N/A	3	33114092	33114093
Gangliosidosis GM1	GLB1	176GtoA | R59H	rs72555392	3	33114105	33114105
Gangliosidosis GM1	GLB1	175CtoT | R59C	N/A	3	33114106	33114106
Gangliosidosis GM1	GLB1	171CtoG | Y57X	N/A	3	33114110	33114110
Leukonychia	PLCD1	1720GtoA | A574T	N/A	3	38050599	38050599
Negative shift activation	SCN5A	3269CtoT | P1090L	N/A	3	38620946	38620946
Sudden infant death syndrome	SCN5A	3250GtoA | G1084S	N/A	3	38620965	38620965
Brugada syndrome	SCN5A	3236CtoA | S1079Y	N/A	3	38620979	38620979
Long QT syndrome	SCN5A	3236CtoT | S1079F	N/A	3	38620979	38620979
Atrial fibrillation	SCN5A	1963GtoA | E655K	N/A	3	38640469	38640469
Long QT syndrome	SCN5A	1960GtoA | E654K	N/A	3	38640472	38640472
Brugada syndrome	SCN5A	1950_1953delAGAT | 1950_1953del4 | 1950_1953del	N/A	3	38640479	38640482
Long QT syndrome	SCN5A	1943CtoT | P648L	rs45609733	3	38640489	38640489
Brugada syndrome	SCN5A	1940CtoA | A647D	rs185638763	3	38640492	38640492
Brugada syndrome	SCN5A	1936delC | 1936del1 | 1936del	N/A	3	38640496	38640496
Congenital cataracts	FYCO1	4127TtoC | L1376P	N/A	3	45972687	45972687
Deafness	TMIE	122_125dupCGCC | 122_125dup4 | 122_125dup	N/A	3	46747311	46747312
Deafness	TMIE	212minus2AtoC	N/A	3	46750614	46750614
Deafness	TMIE	241CtoT | R81C	rs28942096	3	46750645	46750645
Primary failure of tooth eruption	PTH1R	1148GtoA | R383Q	N/A	3	46943287	46943287
Metaphyseal chondrodysplasia	PTH1R	1148GtoA	N/A	3	46943287	46943287
Metaphyseal chondrodysplasia	PTH1R	1228AtoC | T410P	rs121434598	3	46944032	46944032
Metaphyseal chondrodysplasia	PTH1R	1229CtoG | T410R	rs121434602	3	46944033	46944033
Grey platelet syndrome	NBEAL2	2701CtoT | R901X	N/A	3	47038800	47038800
Epidermolysis bullosa dystrophica	COL7A1	8407plus5GtoC	N/A	3	48603695	48603695
Epidermolysis bullosa	COL7A1	8393TtoA | M2798K	rs121912828	3	48603714	48603714

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Epidermolysis bullosa dystrophica	COL7A1	8393TtoG | M2798R	N/A	3	48603714	48603714
Epidermolysis bullosa	COL7A1	8386CtoT | Q2796X	N/A	3	48603721	48603721
Epidermolysis bullosa dystrophica	COL7A1	8375delG | 8375del1 | 8375del	N/A	3	48603732	48603732
Epidermolysis bullosa dystrophica	COL7A1	8371CtoT | R2791W	rs142566193	3	48603736	48603736
Epidermolysis bullosa dystrophica	COL7A1	7756dupC | 7756dup1 | 7756dup	N/A	3	48606218	48606219
Epidermolysis bullosa dystrophica	COL7A1	7741GtoA | G2581R	N/A	3	48606234	48606234
Epidermolysis bullosa dystrophica	COL7A1	7730delC | 7730del1 | 7730del	N/A	3	48606245	48606245
Epidermolysis bullosa dystrophica	COL7A1	7723GtoA | G2575R	N/A	3	48606252	48606252
Epidermolysis bullosa pruriginosa	COL7A1	5097plus1GtoC	N/A	3	48617732	48617732
Epidermolysis bullosa dystrophica	COL7A1	5097GtoA	N/A	3	48617733	48617733
Epidermolysis bullosa dystrophica	COL7A1	5096CtoT | P1699L	rs121912845	3	48617734	48617734
Epidermolysis bullosa	COL7A1	5092delG | 5092del1 | 5092del	N/A	3	48617738	48617738
Epidermolysis bullosa dystrophica	COL7A1	5090GtoA | G1697E	N/A	3	48617740	48617740
Epidermolysis bullosa dystrophica	COL7A1	5087delG | 5087del1 | 5087del	N/A	3	48617743	48617743
Epidermolysis bullosa dystrophica	COL7A1	5086CtoT | R1696C	rs2229820	3	48617744	48617744
Epidermolysis bullosa dystrophica	COL7A1	5081GtoT | G1694V	N/A	3	48617749	48617749
Epidermolysis bullosa dystrophica	COL7A1	5080GtoT | G1694C	N/A	3	48617750	48617750
Epidermolysis bullosa dystrophica	COL7A1	5066CtoG | S1689X	N/A	3	48617764	48617764
Epidermolysis bullosa dystrophica	COL7A1	5017GtoA | G1673R	N/A	3	48618049	48618049
Epidermolysis bullosa dystrophica	COL7A1	5015delA | 5015del1 | 5015del	N/A	3	48618051	48618051
Epidermolysis bullosa dystrophica	COL7A1	5001dupG | 5001dup1 | 5001dup	N/A	3	48618064	48618065
Epidermolysis bullosa dystrophica	COL7A1	5001delG | 5001del1 | 5001del	N/A	3	48618065	48618065
Epidermolysis bullosa dystrophica	COL7A1	4991GtoC | G1664A	N/A	3	48618075	48618075
Epidermolysis bullosa dystrophica	COL7A1	4613GtoA | R1538H	rs2229824	3	48619778	48619778
Epidermolysis bullosa dystrophica	COL7A1	4430delG | 4430del1 | 4430del	N/A	3	48620833	48620833
Epidermolysis bullosa dystrophica	COL7A1	4406_4415delCACGGGGACC | 4406_4415del10 | 4406_4415del	N/A	3	48620848	48620857
Epidermolysis bullosa dystrophica	COL7A1	4351GtoT | E1451X	N/A	3	48621039	48621039
Epidermolysis bullosa dystrophica	COL7A1	3970delC | 3970del1 | 3970del	N/A	3	48622474	48622474
Epidermolysis bullosa dystrophica	COL7A1	3948dupT | 3948dup1 | 3948dup	N/A	3	48622495	48622496
Epidermolysis bullosa dystrophica	COL7A1	3942dupG | 3942dup1 | 3942dup	N/A	3	48622501	48622502
Epidermolysis bullosa dystrophica	COL7A1	151CtoG | R51G	N/A	3	48631916	48631916
Epidermolysis bullosa dystrophica	COL7A1	142TtoC | S48P	N/A	3	48631925	48631925
Epidermolysis bullosa dystrophica	COL7A1	115dupA | 115dup1 | 115dup	N/A	3	48631951	48631952
Epidermolysis bullosa	COL7A1	110_111insA | 110_111ins1 | 110_111ins	N/A	3	48631956	48631957
Carnitine-acylcarnitine carrier deficiency	SLC25A20	691GtoC | D231H	N/A	3	48896552	48896552
Carnitine-acylcarnitine carrier deficiency	SLC25A20	689CtoG | P230R	N/A	3	48896554	48896554
Myocardial infarction, association with	ITIH3	108GtoA | P36P	rs2286797	3	52829633	52829633
Decreased protein stability	ARF4	203TtoC | V68A	rs11550597	3	57569680	57569680
Larsen syndrome, autosomal dominant	FLNB	4292TtoG | L1431R	rs80356511	3	58116537	58116537
Larsen syndrome, autosomal dominant	FLNB	4580TtoA | L1527H	N/A	3	58120408	58120408
Larsen syndrome, autosomal dominant	FLNB	4580TtoC | L1527P	N/A	3	58120408	58120408
Waardenburg syndrome 2	MITF	742_747delAAAGCAinsTAG | 742_747del6ins3 | 742_747delins	N/A	3	70008455	70008460
Waardenburg syndrome	MITF	748TtoC | S250P	rs104893744	3	70008461	70008461
Waardenburg syndrome 2	MITF	758AtoG | Y253C	N/A	3	70008471	70008471
Waardenburg syndrome 2A	MITF	775CtoT | R259X	N/A	3	70008488	70008488
Waardenburg syndrome 2	MITF	806delA | 806del1 | 806del	N/A	3	70008519	70008519
Kallmann syndrome	PROK2	94GtoC | G32R	rs104893767	3	71834110	71834110
Kallmann syndrome	PROK2	70GtoC | A24P	N/A	3	71834134	71834134
Kallmann syndrome	PROK2	NULL	N/A	3	71834207	71834207
Deep vein thrombosis	PROS1	846TtoG | C282W	N/A	3	93617295	93617295
Attenuated ligand activation	NR1I2	365GtoA | R122Q	rs12721608	3	119530419	119530419
Alkaptonuria	HGD	860CtoA | S287X	N/A	3	120360455	120360455
Alkaptonuria	HGD	828GtoC | K276N	N/A	3	120360487	120360487
Alkaptonuria	HGD	533AtoG | E178G	N/A	3	120365836	120365836
Alkaptonuria	HGD	175delA | 175del1 | 175del	N/A	3	120393749	120393749
Alkaptonuria	HGD	171GtoC | K57N	N/A	3	120393753	120393753
Senior-Loken syndrome	IQCB1	1363CtoT | R455X	N/A	3	121500637	121500637
Senior-Loken syndrome	IQCB1	333delT | 333del1 | 333del	N/A	3	121544958	121544958
Hypercalciuria, hypocalcaemic	CASR	1256AtoG | N419S	N/A	3	121981138	121981138
Hypercalcaemia	CASR	1267_1268delGCinsAA | 1267_1268del2ins2 | 1267_1268delins	N/A	3	121981149	121981150
Hypercalcaemia, hypocalciuric	CASR	1370CtoT | A457V	N/A	3	121981252	121981252
Hypercalcaemia, hypocalciuric	CASR	1376AtoG | Q459R	N/A	3	121981258	121981258
Hypercalcaemia, hypocalciuric	CASR	1525GtoA | G509R	N/A	3	121994806	121994806
Hypercalcaemia	CASR	1526GtoC | G509A	N/A	3	121994807	121994807
Hypoparathyroidism	CASR	1541AtoG | Y514C	N/A	3	121994822	121994822
Hypercalcaemia, hypocalciuric	CASR	1555GtoT | E519X	N/A	3	121994836	121994836
Bernard-Soulier syndrome	GP9	70TtoC | C24R	rs28933378	3	128780652	128780652
Bernard-Soulier syndrome	GP9	72TtoG | C24W	N/A	3	128780654	128780654
Bernard-Soulier syndrome	GP9	110AtoG | D37G	rs121918036	3	128780692	128780692
Bernard-Soulier syndrome	GP9	119delG | 119del1 | 119del	N/A	3	128780701	128780701
Hypertrophic osteoarthropathy, primary	SLCO2A1	1105plus4AtoG	N/A	3	133667376	133667376
Hermansky-Pudlak syndrome	HPS3	1189CtoT | R397W	rs121908316	3	148868411	148868411
Aceruloplasminaemia	CP	1282_1286dupTACAC | 1282_1286dup5 | 1282_1286dup	N/A	3	148919950	148919951
Short rib-polydactyly syndrome, type 3	IFT80	721GtoC | G241R	rs138004478	3	160073857	160073857
Cirrhosis	TERC	NULL	N/A	3	169482605	169482605
3-methylcrotonyl-CoA carboxylase deficiency	MCCC1	1380TtoG | I460M	rs119103218	3	182755220	182755220
3-methylcrotonyl-CoA carboxylase deficiency	MCCC1	1193_1194delTG | 1193_1194del2 | 1193_1194del	N/A	3	182759428	182759429
3-methylcrotonylglycinuria	MCCC1	1155AtoC | R385S	rs28934881	3	182759467	182759467
Increased plasma insulin levels, association wi	PARL	784CtoG | L262V	rs3732581	3	183558402	183558402
Idiopathic epilepsy, generalised	CLCN2	2156CtoT | S719L	rs138573287	3	184070586	184070586
Childhood absence epilepsy, association with	CLCN2	2154GtoC | E718D	rs2228292	3	184070588	184070588
Hypomagnesemia with hypercalciuria and nep	CLDN16	485GtoT | G162V	N/A	3	190122608	190122608
Hypomagnesemia with hypercalciuria and nep	CLDN16	547AtoG | K183E	N/A	3	190122670	190122670
Optic atrophy & spastic paraplegia	OPA1	876_878delTGT | 876_878del3 | 876_878del	N/A	3	193355746	193355748
Optic atrophy 1	OPA1	878_883delTGGTTG | 878_883del6 | 878_883del	N/A	3	193355748	193355753
Mucopolysaccharidosis I	IDUA	NULL	N/A	4	980871	980871
Hurler syndrome	IDUA	1AtoC | M1L	N/A	4	980873	980873
Mucopolysaccharidosis I	IDUA	2TtoC | M1T	N/A	4	980874	980874
Hurler syndrome	IDUA	46_57delTCGCTCCTGGCC | 46_57del12 | 46_57del	N/A	4	980918	980929
Hurler syndrome	IDUA	65delC | 65del1 | 65del	N/A	4	980937	980937
Mucopolysaccharidosis I	IDUA	98AtoC | H33P	N/A	4	980970	980970
Hurler syndrome	IDUA	1190minus1GtoA	N/A	4	996519	996519
Hurler syndrome	IDUA	1190-1delG | 1190-1del1 | 1190-1del	N/A	4	996519	996519
Scheie syndrome	IDUA	1198CtoT | Q400X	N/A	4	996528	996528
Hurler syndrome	IDUA	1205GtoA | W402X	rs121965019	4	996535	996535

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Hurler syndrome	IDUA	1210GtoT | E404X	N/A	4	996540	996540
Hurler syndrome	IDUA	1225GtoC | G409R	rs11934801	4	996555	996555
Mucopolysaccharidosis I	IDUA	1227dupG | 1227dup1 | 1227dup	N/A	4	996557	996558
Hurler syndrome	IDUA	1603CtoT | L535F	N/A	4	997211	997211
Hurler syndrome	IDUA	1614delG | 1614del1 | 1614del	N/A	4	997222	997222
Hypoacetylaspartia	NAT8L	212_230delGGCCGCCGGGGGGGCGCGG | 212_230del19 | 212_230del	N/A	4	2061450	2061468
Congenital myasthenic syndromes	DOK7	7GtoA | E3K	N/A	4	3465109	3465109
Congenital myasthenic syndromes	DOK7	48CtoT	N/A	4	3465150	3465150
Congenital myasthenic syndromes	DOK7	55minus2AtoC	N/A	4	3465231	3465231
Congenital myasthenic syndromes	DOK7	91CtoA | P31T	N/A	4	3465269	3465269
Congenital myasthenic syndromes	DOK7	98CtoT | A33V	N/A	4	3465276	3465276
Congenital myasthenic syndromes	DOK7	101minus1GtoT	N/A	4	3475132	3475132
Congenital myasthenic syndromes	DOK7	134CtoT | S45L	rs62272670	4	3475166	3475166
Congenital myasthenic syndromes	DOK7	190GtoA | G64R	N/A	4	3475222	3475222
Congenital myasthenic syndromes	DOK7	230CtoT | T77M	N/A	4	3475262	3475262
Congenital myasthenic syndromes	DOK7	325GtoT | G109C	N/A	4	3475357	3475357
Congenital myasthenic syndromes	DOK7	331plus1GtoT	N/A	4	3475364	3475364
Impaired coupling, association with	ADRA2C	971_982delGGCCGGGGGCGG | 971_982del12 | 971_982del	N/A	4	3769304	3769315
Ellis-van Creveld syndrome	EVC	617GtoA | S206N	N/A	4	5733384	5733384
Ellis-van Creveld syndrome	EVC	2200CtoT | Q734X	N/A	4	5800415	5800415
Wolfram syndrome	WFS1	376GtoA | A126T	rs145639028	4	6290774	6290774
Wolfram syndrome	WFS1	387GtoA | W129X	N/A	4	6290785	6290785
Wolfram syndrome	WFS1	397GtoA | A133T	N/A	4	6290795	6290795
Wolfram syndrome	WFS1	406CtoT | Q136X	N/A	4	6290804	6290804
Wolfram syndrome	WFS1	409_424dupGGCCGTCGCGAGGCTG | 409_424dup16 | 409_424dup	N/A	4	6290822	6290823
Wolfram syndrome	WFS1	460plus1GtoA	N/A	4	6290859	6290859
Wolfram syndrome	WFS1	639_642delGGCG | 639_642del4 | 639_642del	N/A	4	6293651	6293654
Wolfram syndrome	WFS1	670CtoT | Q224X	N/A	4	6293682	6293682
Wolfram syndrome	WFS1	676CtoT | Q226X	rs104893880	4	6293688	6293688
Renal hypouricaemia	SLC2A9	224TtoG | L75R	N/A	4	10020624	10020624
Meckel syndrome	CC2D2A	1762CtoT	N/A	4	15538697	15538697
Cone-rod dystrophy	PROM1	1354dupT | 1354dup1 | 1354dup	N/A	4	16008260	16008261
Dihydropteridine reductase deficiency	QDPR	515CtoT | P172L	N/A	4	17493885	17493885
Dihydropteridine reductase deficiency	QDPR	508GtoA | G170S	N/A	4	17493892	17493892
Dihydropteridine reductase deficiency	QDPR	472CtoT | H158Y	N/A	4	17493928	17493928
Parkinson disease, lower risk, association with	UCHL1	53CtoA | S18Y	rs5030732	4	41259633	41259633
Central hypoventilation syndrome	PHOX2B	618dupC | 618dup1 | 618dup	N/A	4	41748150	41748151
Central hypoventilation syndrome	PHOX2B	618delC | 618del1 | 618del	N/A	4	41748151	41748151
Central hypoventilation syndrome	PHOX2B	617_618insT | 617_618ins1 | 617_618ins	N/A	4	41748151	41748152
Central hypoventilation syndrome	PHOX2B	616_617insT | 616_617ins1 | 616_617ins	N/A	4	41748152	41748153
Neuroblastoma	PHOX2B	600delC | 600del1 | 600del	N/A	4	41748169	41748169
Central hypoventilation syndrome	PHOX2B	590dupG | 590dup1 | 590dup	N/A	4	41748178	41748179
Neuroblastoma	PHOX2B	590GtoA | G197D	rs104893856	4	41748179	41748179
Central hypoventilation syndrome	PHOX2B	577_583delGACAGCAinsCCCCGCAC | 577_583del7ins8 | 577_583delins	N/A	4	41748186	41748192
Central hypoventilation syndrome	PHOX2B	577delG | 577del1 | 577del	N/A	4	41748192	41748192
Central hypoventilation syndrome	PHOX2B	575delC | 575del1 | 575del	N/A	4	41748194	41748194
Alpha-fetoprotein deficiency	AFP	883_884delCT | 883_884del2 | 883_884del	N/A	4	74313218	74313219
Fraser syndrome	FRAS1	3877CtoT | H1293Y	N/A	4	79308757	79308757
Fibromatosis, juvenile hyaline	ANTXR2	876_877insCAA | 876_877ins3 | 876_877ins	N/A	4	80940120	80940121
Coenzyme Q10 deficiency	COQ2	437GtoA | S146N	rs121918233	4	84200234	84200234
Dentinogenesis imperfecta type II	DSPP	44CtoT | A15V	rs121912989	4	88532104	88532104
Dentinogenesis imperfecta type I	DSPP	49CtoA | P17T	rs121912986	4	88532109	88532109
Dentinogenesis imperfecta type II	DSPP	49CtoT | P17S	N/A	4	88532109	88532109
Dentinogenesis imperfecta type II	DSPP	50CtoT | P17L	N/A	4	88532110	88532110
Polycystic kidney disease 2	PKD2	469CtoT | R157X	N/A	4	88929354	88929354
Polycystic kidney disease 2	PKD2	478CtoT | Q160X	N/A	4	88929363	88929363
Polycystic kidney disease 2	PKD2	538dupC | 538dup1 | 538dup	N/A	4	88929423	88929424
Polycystic kidney disease 2	PKD2	556CtoT | R186X	N/A	4	88929441	88929441
Polycystic kidney disease 2	PKD2	567GtoA | W189X	N/A	4	88929452	88929452
Polycystic kidney disease 2	PKD2	568GtoA | A190T	N/A	4	88929453	88929453
Mannosidosis, beta, lysosomal	MANBA	247GtoT | E83X	rs121434335	4	103647771	103647771
Dysfibrinogenaemia	FGB	211TtoA | Y71N	N/A	4	155487056	155487056
Hypofibrinogenaemia	FGB	213TtoG | Y71X	N/A	4	155487058	155487058
Dysfibrinogenaemia	FGB	220CtoT | R74C	rs121909619	4	155487065	155487065
Afibrinogenaemia	FGB	248_249delAGinsT | 248_249del2ins1 | 248_249delins	N/A	4	155487093	155487094
Hypofibrinogenaemia	FGB	264delA | 264del1 | 264del	N/A	4	155487109	155487109
Mitochondrial myopathy & hypertrophic cardi	SLC25A4	111plus1GtoA	N/A	4	186064638	186064638
Factor XI deficiency	F11	569TtoC | L190P	N/A	4	187197024	187197024
Factor XI deficiency	F11	1304plus12GtoA	rs116667976	4	187205426	187205426
Factor XI deficiency	F11	1760GtoC | W587S	rs121965072	4	187209650	187209650
Factor XI deficiency	F11	1778CtoT | T593M	rs145906668	4	187209668	187209668
Factor XI deficiency	F11	1782CtoA | S594R	rs28934609	4	187209672	187209672
Factor XI deficiency	F11	1786GtoT | G596C	N/A	4	187209676	187209676
Factor XI deficiency	F11	1789GtoA | E597K	N/A	4	187209679	187209679
Factor XI deficiency	F11	1797TtoA | C599X	N/A	4	187209687	187209687
Hartnup disorder	SLC6A19	1787_1788insG | 1787_1788ins1 | 1787_1788ins	N/A	5	1221901	1221902
Aplastic anaemia	TERT	2080GtoA | V694M	rs121918662	5	1279456	1279456
Pulmonary fibrosis, idiopathic	TERT	2011CtoT | R671W	N/A	5	1279525	1279525
Dystonia-parkinsonism, infantile	SLC6A3	1103TtoA | L368Q	N/A	5	1414859	1414859
Mutilating sensory neuropathy with spastic pa	CCT5	440AtoG | H147R	rs118203986	5	10256175	10256175
Multiple sclerosis, increased risk, association v	IL7R	731TtoC | I244T	rs6897932	5	35874575	35874575
Cornelia de Lange syndrome	NIPBL	1513dupA | 1513dup1 | 1513dup	N/A	5	36984795	36984796
Cornelia de Lange syndrome	NIPBL	1547dupG | 1547dup1 | 1547dup	N/A	5	36984829	36984830
Cornelia de Lange syndrome	NIPBL	1548delT | 1548del1 | 1548del	N/A	5	36984830	36984830
Cornelia de Lange syndrome	NIPBL	5918AtoC | Q1973P	N/A	5	37036536	37036536
Cornelia de Lange syndrome	NIPBL	6108plus2TtoC	N/A	5	37038842	37038842
Glucocorticoid deficiency	NNT	1310CtoT | P437L	N/A	5	43645478	43645478
Glucocorticoid deficiency	NNT	1355delA | 1355del1 | 1355del	N/A	5	43645523	43645523
Mucopolysaccharidosis VI	ARSB	1079TtoC | L360P	N/A	5	78181470	78181470
Reduced activity	ARSB	1072GtoA | V358M	N/A	5	78181477	78181477
Mucopolysaccharidosis VI	ARSB	750_754delATACTinsCCTGAAGTCAAG | 750_754del5ins12 | 750_754delins	N/A	5	78251262	78251266
Mucopolysaccharidosis VI	ARSB	753CtoG | Y251X	N/A	5	78251263	78251263
Colon cancer, association with	MSH3	235GtoA | V79I	rs1650697	5	79950781	79950781
Obesity	PCSK1	375GtoA | M125I	rs146545244	5	95761545	95761545
Adenomatous polyposis coli	APC	1787CtoG | S596X	N/A	5	112170691	112170691

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Adenomatous polyposis coli	APC	1797CtoA | C599X	N/A	5	112170701	112170701
Adenomatous polyposis coli	APC	1812delT | 1812del1 | 1812del	N/A	5	112170716	112170716
Adenomatous polyposis coli	APC	1816dupA | 1816dup1 | 1816dup	N/A	5	112170720	112170721
Adenomatous polyposis coli	APC	1817delT | 1817del1 | 1817del	N/A	5	112170721	112170721
Adenomatous polyposis coli	APC	1818dupA | 1818dup1 | 1818dup	N/A	5	112170722	112170723
Adenomatous polyposis coli	APC	1822_1823insA | 1822_1823ins1 | 1822_1823ins	N/A	5	112170726	112170727
Adenomatous polyposis coli	APC	1823_1824insA | 1823_1824ins1 | 1823_1824ins	N/A	5	112170727	112170728
Adenomatous polyposis coli	APC	1825delG | 1825del1 | 1825del	N/A	5	112170729	112170729
Adenomatous polyposis coli	APC	1829delA | 1829del1 | 1829del	N/A	5	112170733	112170733
Adenomatous polyposis coli	APC	1840dupG | 1840dup1 | 1840dup	N/A	5	112170744	112170745
Adenomatous polyposis coli	APC	1938_1939delAGinsC | 1938_1939del2ins1 | 1938_1939delins	N/A	5	112170842	112170843
Adenomatous polyposis coli	APC	1946dupA | 1946dup1 | 1946dup	N/A	5	112170850	112170851
Adenomatous polyposis coli	APC	1946_1947insC | 1946_1947ins1 | 1946_1947ins	N/A	5	112170850	112170851
Adenomatous polyposis coli	APC	1948GtoT | E650X	N/A	5	112170852	112170852
Adenomatous polyposis coli	APC	1954_1958+15delCACAGGTATATATAGAGTTT | 1954_1958+15del20 | 1954_1958+15del	N/A	5	112170858	112170877
Adenomatous polyposis coli	APC	1956CtoT	N/A	5	112170860	112170860
Adenomatous polyposis coli	APC	1957AtoC	N/A	5	112170861	112170861
Adenomatous polyposis coli	APC	1957AtoG	N/A	5	112170861	112170861
Adenomatous polyposis coli	APC	1957AtoT	N/A	5	112170861	112170861
Colorectal adenoma	APC	1958GtoA | R653K	N/A	5	112170862	112170862
Adenomatous polyposis coli	APC	1958GtoA	N/A	5	112170862	112170862
Adenomatous polyposis coli	APC	1958GtoC	N/A	5	112170862	112170862
Adenomatous polyposis coli	APC	1958GtoT	N/A	5	112170862	112170862
Adenomatous polyposis coli	APC	1958plus1GtoA	N/A	5	112170863	112170863
Adenomatous polyposis coli	APC	1958plus1GtoC	N/A	5	112170863	112170863
Adenomatous polyposis coli	APC	1958plus1GtoT	N/A	5	112170863	112170863
Adenomatous polyposis coli	APC	1958+1delG | 1958+1del1 | 1958+1del	N/A	5	112170863	112170863
Adenomatous polyposis coli	APC	1958plus2TtoC	N/A	5	112170864	112170864
Adenomatous polyposis coli	APC	1958plus2TtoG	N/A	5	112170864	112170864
Adenomatous polyposis coli	APC	1958+1_1958+2dupGT | 1958+1_1958+2dup2 | 1958+1_1958+2dup	N/A	5	112170864	112170865
Adenomatous polyposis coli	APC	1958plus3AtoC	N/A	5	112170865	112170865
Adenomatous polyposis coli	APC	1958plus3AtoG	N/A	5	112170865	112170865
Epilepsy, pyridoxine-dependent	ALDH7A1	848GtoA | G283D	N/A	5	125903974	125903974
Epilepsy, pyridoxine-dependent	ALDH7A1	842delA | 842del1 | 842del	N/A	5	125903980	125903980
Alpha-AASA dehydrogenase deficiency	ALDH7A1	834GtoA	N/A	5	125903988	125903988
Epilepsy, pyridoxine-dependent	ALDH7A1	833delT | 833del1 | 833del	N/A	5	125903989	125903989
Pseudohypoaldosteronism type II	KLHL3	232AtoG | M78V	N/A	5	137045448	137045448
Pseudohypoaldosteronism type II	KLHL3	230CtoA | A77E	N/A	5	137045450	137045450
Parkinson disease	HSPA9	1525CtoT | P509S	N/A	5	137893666	137893666
Increased glucocorticoid sensitivity	NR3C1	1201GtoC | D401H	N/A	5	142693717	142693717
Infantile hepatopathy	LARS	1118AtoG | Y373C	N/A	5	145533491	145533491
Pancreatitis, chronic	SPINK1	87plus26TtoC	N/A	5	147209136	147209136
Pancreatitis, chronic	SPINK1	87plus1GtoA	N/A	5	147209161	147209161
Charcot-Marie-Tooth disease 1	SH3TC2	505TtoC | Y169H	rs80359890	5	148422281	148422281
Charcot-Marie-Tooth disease 4C	SH3TC2	253delG | 253del1 | 253del	N/A	5	148427451	148427451
Charcot-Marie-Tooth disease 4C	SH3TC2	217_227delGCTGCTCGGAGinsCCAGTAA | 217_227del11ins7 | 217_227delins	rs80338919	5	148427477	148427487
Charcot-Marie-Tooth disease 4C	SH3TC2	28delG | 28del1 | 28del	rs80338918	5	148442558	148442558
Treacher-Collins syndrome	TCOF1	514_515delGA | 514_515del2 | 514_515del	N/A	5	149748414	149748415
Treacher-Collins syndrome	TCOF1	827_844delGATCTGAAAGTGAGGAGG | 827_844del18 | 827_844del	N/A	5	149751756	149751773
Treacher-Collins syndrome	TCOF1	2426delC | 2426del1 | 2426del	N/A	5	149758553	149758553
Muscular dystrophy, limb girdle	SGCD	631AtoT | N211Y	N/A	5	156184647	156184647
Muscular dystrophy, limb girdle	SGCD	657delC | 657del1 | 657del	N/A	5	156184673	156184673
Asthma severity in males, association with	KCNMB1	418CtoT | R140W	rs76348543	5	169805866	169805866
Hypertension, response to verapamil, associat	KCNMB1	328GtoC | V110L	rs2301149	5	169805956	169805956
Frank-Ter Haar syndrome	SH3PXD2B	147dupT | 147dup1 | 147dup	N/A	5	171849428	171849429
Frank-Ter Haar syndrome	SH3PXD2B	127CtoT | R43W	N/A	5	171849449	171849449
Dementia with Lewy bodies	SNCB	368CtoA | P123H	rs104893937	5	176048219	176048219
Sotos syndrome	NSD1	3796plus33AtoT	N/A	5	176639229	176639229
Sotos syndrome	NSD1	3886AtoT | K1296X	N/A	5	176662911	176662911
Sotos syndrome	NSD1	4417CtoT | R1473X	N/A	5	176673717	176673717
Sotos syndrome	NSD1	5907dupT | 5907dup1 | 5907dup	N/A	5	176709480	176709481
Sotos syndrome	NSD1	5911delT | 5911del1 | 5911del	N/A	5	176709484	176709484
Sotos syndrome	NSD1	5927_5928delTA | 5927_5928del2 | 5927_5928del	N/A	5	176709500	176709501
Sotos syndrome	NSD1	6383dupG | 6383dup1 | 6383dup	N/A	5	176719079	176719080
Night blindness, congenital	GRM6	172GtoC | G58R	rs62638198	5	178421774	178421774
Night blindness, congenital	GRM6	137CtoT | P46L	rs62638197	5	178421809	178421809
Venous thromboembolism and ischaemic stro	LTC4S	374GtoA | G125D	N/A	5	179223316	179223316
Venous thromboembolism and ischaemic stro	LTC4S	451_453+10delTGAGACCAAGGCC | 451_453+10del13 | 451_453+10del	N/A	5	179223393	179223405
Factor XIII deficiency	F13A1	1261GtoA | G421S	N/A	6	6196074	6196074
Factor XIII deficiency	F13A1	1243GtoT | V415F	rs121913070	6	6196092	6196092
Factor XIII deficiency	F13A1	1241CtoG | S414W	N/A	6	6196094	6196094
Factor XIII deficiency	F13A1	1241CtoT | S414L	N/A	6	6196094	6196094
Branchio-oculo-facial syndrome	TFAP2A	724GtoA | E242K	rs151344526	6	10404781	10404781
Branchio-oculo-facial syndrome	TFAP2A	715TtoC | S239P	N/A	6	10404790	10404790
Haemochromatosis, association with	HFE	471delA | 471del1 | 471del	N/A	6	26091672	26091672
Haemochromatosis	HFE	480delC | 480del1 | 480del	N/A	6	26091681	26091681
Haemochromatosis	HFE	502GtoC | E168Q	rs146519482	6	26091703	26091703
Haemochromatosis	HFE	502GtoT | E168X	N/A	6	26091703	26091703
Haemochromatosis	HFE	506GtoA | W169X	N/A	6	26091707	26091707
Haemochromatosis	HFE	527CtoT | A176V	N/A	6	26091728	26091728
Haemochromatosis	HFE	548TtoC | L183P	N/A	6	26091749	26091749
Temporal lobe epilepsy, association with	GABBR1	1465GtoA | G489S	rs144853554	6	29581121	29581121
Peeling skin disease	CDSN	175AtoT | K59X	N/A	6	31085217	31085217
Peeling skin disease	CDSN	164_167dupGCCT | 164_167dup4 | 164_167dup	N/A	6	31085224	31085225
Altered expression	HLA-C	NULL	rs1071643	6	31236639	31236639
Null allele	MICB	205delC | 205del1 | 205del	rs45459705	6	31473528	31473528
Haemolytic uraemic syndrome, atypical	CFB	724AtoC | I242L	rs144812066	6	31915584	31915584
Graves disease, association with	HLA-DRB1	307_308delGCinsCG | 307_308del2ins2 | 307_308delins	N/A	6	32551948	32551949
HLA class I deficiency	TAP2	1867CtoT | R623X	N/A	6	32797242	32797242
Ossification, protection against, association wi	COL11A2	877minus4TtoA	rs138085890	6	33152835	33152835
Reduced agonist responsiveness	GLP1R	20CtoT | P7L	rs10305420	6	39016636	39016636
Reduced agonist responsiveness	GLP1R	59GtoA | R20K	rs10305421	6	39016675	39016675
Reduced agonist responsiveness	GLP1R	1262GtoA | R421Q	rs10305510	6	39053719	39053719
Molybdenum cofactor deficiency	MOCS1	218GtoA | R73Q	N/A	6	39895100	39895100
Molybdenum cofactor deficiency	MOCS1	217CtoT | R73W	rs104893970	6	39895101	39895101

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Molybdenum cofactor deficiency	MOCS1	196_213delGGCCGGCAGCACAGCTAC | 196_213del18 | 196_213del	N/A	6	39895105	39895122
Molybdenum cofactor deficiency	MOCS1	199CtoT | R67W	N/A	6	39895119	39895119
Presenile dementia with bone cysts	TREM2	401AtoG | D134G	rs28939079	6	41127611	41127611
Dementia, early-onset, without bone cysts	TREM2	40+4_40+6delGGA | 40+4_40+6del3 | 40+4_40+6del	N/A	6	41130775	41130777
Presenile dementia with bone cysts	TREM2	40GtoT | E14X	N/A	6	41130781	41130781
Zellweger syndrome	PEX6	311delG | 311del1 | 311del	rs61753209	6	42946578	42946578
Peroxisome biogenesis disorder	PEX6	281CtoA | A94E	rs61752143	6	42946608	42946608
Zellweger syndrome C	PEX6	275_280delTGCGGG | 275_280del6 | 275_280del	rs61752142	6	42946609	42946614
Zellweger syndrome	PEX6	277CtoG | R93G	N/A	6	42946612	42946612
Zellweger syndrome	PEX6	224dupT | 224dup1 | 224dup	N/A	6	42946664	42946665
Peroxisome biogenesis disorder	PEX6	170TtoC | L57P	rs61752140	6	42946719	42946719
Zellweger syndrome	PEX6	115_120delGCCCTG | 115_120del6 | 115_120del	N/A	6	42946769	42946774
Peroxisome biogenesis disorder	PEX6	35TtoC | F12S	rs61752139	6	42946854	42946854
3-M syndrome	CUL7	3608dupG | 3608dup1 | 3608dup	N/A	6	43010576	43010577
Cleidocranial dysplasia	RUNX2	694CtoT | Q232X	N/A	6	45459686	45459686
Cleidocranial dysplasia	RUNX2	716delC | 716del1 | 716del	N/A	6	45459708	45459708
Cleidocranial dysplasia	RUNX2	718_721delAGTT | 718_721del4 | 718_721del	N/A	6	45459710	45459713
Rh-deficiency syndrome	RHAG	236GtoA | S79N	rs121918586	6	49586997	49586997
Stomatocytosis, overhydrated	RHAG	194TtoC | F65S	N/A	6	49587039	49587039
Stomatocytosis, overhydrated	RHAG	182TtoG | I61R	N/A	6	49587051	49587051
Polycystic kidney disease	PKHD1	8737GtoA | V2913M	N/A	6	51619642	51619642
Polycystic kidney disease	PKHD1	6996plus4AtoT	N/A	6	51768391	51768391
Polycystic kidney disease	PKHD1	6418AtoC | T2140P	N/A	6	51776669	51776669
Polycystic kidney disease	PKHD1	6401TtoC | L2134P	N/A	6	51776686	51776686
Polycystic kidney disease	PKHD1	5237minus1GtoA	N/A	6	51887743	51887743
Polycystic kidney disease	PKHD1	4457CtoT | P1486L	N/A	6	51890151	51890151
Polycystic kidney disease	PKHD1	1830TtoA | Y610X	N/A	6	51920391	51920391
Polycystic kidney disease	PKHD1	1774CtoT | R592X	N/A	6	51920447	51920447
Polycystic kidney disease	PKHD1	1751GtoT | G584V	N/A	6	51920470	51920470
Polycystic kidney disease	PKHD1	976plus2TtoA	N/A	6	51929751	51929751
Polycystic kidney disease	PKHD1	920TtoC | I307T	N/A	6	51929809	51929809
Polycystic kidney disease	PKHD1	881minus1GtoA	N/A	6	51929849	51929849
Autonomic sensory neuropathy	dst1eA	13887delA | 13887del1 | 13887del	N/A	6	56341055	56341055
Maple syrup urine disease	BCKDHB	163_166dupACTT | 163_166dup4 | 163_166dup	N/A	6	80816576	80816577
Pontocerebellar hypoplasia	RARS2	110plus5AtoG	rs63749985	6	88279230	88279230
Polycystic liver disease	SEC63	2006_2007delAT | 2006_2007del2 | 2006_2007del	N/A	6	108197795	108197796
Oculodentodigital dysplasia	GJA1	578CtoT | P193L	N/A	6	121768571	121768571
Oculodentodigital dysplasia	GJA1	581AtoC | H194P	rs104893966	6	121768574	121768574
Oculodentodigital dysplasia	GJA1	646GtoT | V216L	N/A	6	121768639	121768639
Heart malformations	GJA1	1127GtoA | R376Q	rs104893965	6	121769120	121769120
Muscular dystrophy, merosin deficient	LAMA2	4524minus2AtoG	N/A	6	129674307	129674307
Muscular dystrophy, merosin deficient	LAMA2	4524minus2AtoT	N/A	6	129674307	129674307
Arginase deficiency	ARG1	365GtoA | W122X	rs104893947	6	131902418	131902418
Hyperargininaemia	ARG1	383AtoG | D128G	rs140549609	6	131902436	131902436
Arginase deficiency	ARG1	386_388delTCA | 386_388del3 | 386_388del	N/A	6	131902439	131902441
Arginase deficiency	ARG1	413GtoT | G138V	rs104893943	6	131902466	131902466
Hyperargininaemia	ARG1	422AtoT | H141L	N/A	6	131902475	131902475
Argininaemia	ARG1	425GtoA | G142E	N/A	6	131902478	131902478
Refsum disease	PEX7	13_19dupTGCGGTG | 13_19dup7 | 13_19dup	N/A	6	137143822	137143823
Refsum disease	PEX7	40AtoC | T14P	rs61753233	6	137143843	137143843
Rhizomelic chondrodysplasia punctata	PEX7	45_52delGGGACGCC | 45_52del8 | 45_52del	N/A	6	137143848	137143855
Rhizomelic chondrodysplasia punctata	PEX7	45_52dupGGGACGCC | 45_52dup8 | 45_52dup	N/A	6	137143855	137143856
Epilepsy, progressive myoclonus	EPM2A	94TtoG | W32G	rs104893955	6	146056541	146056541
Epilepsy, progressive myoclonus	EPM2A	82GtoA | E28K	N/A	6	146056553	146056553
Epilepsy, progressive myoclonus	EPM2A	73TtoC | S25P	N/A	6	146056562	146056562
Decreased receptor expression	OPRM1	570TtoA | N190K	rs34074916	6	154411240	154411240
Intellectual disability	ARID1B	1114dupC | 1114dup1 | 1114dup	N/A	6	157100177	157100178
Reduced metformin uptake	SLC22A1	566CtoT | S189L	rs34104736	6	160553314	160553314
Altered substrate specificity	SLC22A3	1199CtoT | T400I	rs8187725	6	160858154	160858154
Lp(a) deficiency	LPA	5053TtoC | W1685R	N/A	6	160969612	160969612
Plasminogen deficiency	PLG	974GtoA | R325H	N/A	6	161139748	161139748
Plasminogen deficiency	PLG	977AtoT | N326I	N/A	6	161139751	161139751
Plasminogen deficiency	PLG	1016_1018dupACA | 1016_1018dup3 | 1016_1018dup	N/A	6	161139792	161139793
Dental developmental defects	SMOC2	84plus1GtoT	N/A	6	168842135	168842135
Walker-Warburg syndrome	ISPD	53dupT | 53dup1 | 53dup	N/A	7	16460894	16460895
Mulleran duct anomaly	HOXA10	170AtoG | Y57C	N/A	7	27213756	27213756
Hand-foot-genital syndrome	HOXA13	407CtoA | S136X	N/A	7	27239290	27239290
Hand-foot-genital syndrome	HOXA13	234_251dupTGCGGCGGCGGCCGCGGC | 234_251dup18 | 234_251dup	N/A	7	27239445	27239446
Reduced promoter activity	NPSR1	NULL	rs2530547	7	34697922	34697922
Trichothiodystrophy, nonphotosensitive	C7orf11	277delT | 277del1 | 277del	N/A	7	40173890	40173890
Cerebral cavernous malformations	CCM2	30GtoA	N/A	7	45039962	45039962
Cerebral cavernous malformations	CCM2	30plus1GtoA	N/A	7	45039963	45039963
Cerebral cavernous malformations	CCM2	30+5_30+6delGCinsTT | 30+5_30+6del2ins2 | 30+5_30+6delins	N/A	7	45039967	45039968
Cerebral cavernous malformations	CCM2	353dupA | 353dup1 | 353dup	N/A	7	45104126	45104127
Phenotype modifier, association with	CCM2	358GtoA | V120I	rs11552377	7	45104131	45104131
Teratozoospermia	ZPBP	50GtoC | R17P	N/A	7	50132741	50132741
Aromatic L-amino acid decarboxylase deficien	DDC	925TtoC | F309L	rs137853209	7	50563067	50563067
Glioblastoma, risk, association with	EGFR	NULL	rs712829	7	55086755	55086755
3-phosphoserine phosphatase deficiency	PSPH	155TtoC | M52T	rs104894036	7	56087413	56087413
Argininosuccinate lyase deficiency	ASL	637CtoT | R213X	N/A	7	65552355	65552355
Altered enzyme activity	POR	1414GtoA | V472M	rs72557946	7	75614912	75614912
Adrenal hyperplasia	POR	1415_1416delTG | 1415_1416del2 | 1415_1416del	N/A	7	75614913	75614914
Adrenal hyperplasia	POR	1493GtoC | R498P	N/A	7	75614991	75614991
Altered enzyme activity	POR	1508CtoT | A503V	rs1057868	7	75615006	75615006
Disordered steroidogenesis	POR	1535_1551dupTGCCCATGTTCGTGCGC | 1535_1551dup17 | 1535_1551dup	N/A	7	75615049	75615050
Disordered steroidogenesis	POR	1615GtoA | G539R	rs121912976	7	75615113	75615113
Neuropathy, distal hereditary motor	HSPB1	421AtoC | K141Q	N/A	7	75933175	75933175
Altered transcription factor binding	FZD1	NULL	rs2232157	7	90893972	90893972
Osteogenesis imperfecta II/III	COL1A2	1252GtoA | G418S	rs72658110	7	94040368	94040368
Osteogenesis imperfecta II/III	COL1A2	1253GtoA | G418D	rs72658111	7	94040369	94040369
Osteogenesis imperfecta IV	COL1A2	1270GtoA | G424S	rs72658112	7	94040386	94040386
Osteogenesis imperfecta III/IV	COL1A2	1279GtoA | G427S	rs72658113	7	94040395	94040395
Osteogenesis imperfecta II	COL1A2	1298GtoA | G433E	rs72658114	7	94040414	94040414
Osteogenesis imperfecta I	COL1A2	2197GtoT | G733C	rs72658172	7	94049862	94049862
Osteogenesis imperfecta I	COL1A2	2206GtoT | G736C	rs72658173	7	94049871	94049871

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Osteogenesis imperfecta III	COL1A2	2215GtoA | G739R	rs72658174	7	94049880	94049880
Osteogenesis imperfecta II	COL1A2	2215GtoC | G739R	N/A	7	94049880	94049880
Osteogenesis imperfecta I	COL1A2	2710GtoC | G904R	rs72659307	7	94054465	94054465
Osteogenesis imperfecta II	COL1A2	2712_2729delGGCCCGTGGTCCTCCTGG | 2712_2729del18 | 2712_2729del	N/A	7	94054467	94054484
Osteogenesis imperfecta IV	COL1A2	2835plus1GtoA	rs72659310	7	94054976	94054976
Citrullinaemia, adult onset, type II	SLC25A13	46GtoT | E16X	N/A	7	95926233	95926233
Citrullinaemia, adult onset, type II	SLC25A13	16minus1GtoA	N/A	7	95926264	95926264
Phenotype modifier	TFR2	1364GtoA | R455Q	rs41303501	7	100226902	100226902
Haemochromatosis	TFR2	1235_1237delACA | 1235_1237del3 | 1235_1237del	N/A	7	100228545	100228547
Haemochromatosis	TFR2	1186CtoT | R396X	rs80338882	7	100228596	100228596
Enlarged vestibular aqueduct & Mondini dysp	SLC26A4	minus3minus2AtoG	N/A	7	107302082	107302082
Deafness, non-syndromic, autosomal recessive	SLC26A4	2TtoC | M1T	rs111033302	7	107302088	107302088
Enlarged vestibular aqueduct	SLC26A4	2TtoG | M1R	N/A	7	107302088	107302088
Increased ion transport activity	SLC26A4	262GtoA | V88I	N/A	7	107303838	107303838
Deafness, non-syndromic, autosomal recessive	SLC26A4	439AtoG | M147V	N/A	7	107314632	107314632
Enlarged vestibular aqueduct	SLC26A4	440TtoC | M147T	N/A	7	107314633	107314633
Enlarged vestibular aqueduct	SLC26A4	441GtoA | M147I	N/A	7	107314634	107314634
Hearing loss	SLC26A4	1102GtoT | G368X	N/A	7	107329598	107329598
Deafness, non-syndromic, autosomal recessive	SLC26A4	1105AtoG | K369E	rs121908361	7	107329601	107329601
Diarrhoea, congenital chloride	SLC26A3	951_953delGGT | 951_953del3 | 951_953del	N/A	7	107427290	107427292
Developmental verbal dyspraxia	FOXP2	50AtoT | Q17L	N/A	7	114066616	114066616
Cystic fibrosis	CFTR	3876delA | 3876del1 | 3876del	N/A	7	117292898	117292898
Cystic fibrosis	CFTR	3877GtoA | V1293I	N/A	7	117292899	117292899
Cystic fibrosis	CFTR	3883_3886delATTT | 3883_3886del4 | 3883_3886del	N/A	7	117292905	117292908
Cystic fibrosis	CFTR	3883delA | 3883del1 | 3883del	N/A	7	117292905	117292905
Cystic fibrosis	CFTR	3883_3884insG | 3883_3884ins1 | 3883_3884ins	N/A	7	117292905	117292906
Cystic fibrosis	CFTR	3889dupT | 3889dup1 | 3889dup	N/A	7	117292911	117292912
Cystic fibrosis	CFTR	3896CtoT | T1299I	N/A	7	117292918	117292918
Cystic fibrosis	CFTR	3898TtoC | F1300L	N/A	7	117292920	117292920
Cystic fibrosis	CFTR	3904AtoT | K1302X	N/A	7	117292926	117292926
Cystic fibrosis	CFTR	3907AtoC | N1303H	rs121909042	7	117292929	117292929
Cystic fibrosis	CFTR	3908AtoT | N1303I	N/A	7	117292930	117292930
Cystic fibrosis	CFTR	3908delA | 3908del1 | 3908del	N/A	7	117292930	117292930
Cystic fibrosis	CFTR	3908dupA | 3908dup1 | 3908dup	N/A	7	117292930	117292931
Cystic fibrosis	CFTR	3909CtoG | N1303K	rs80034486	7	117292931	117292931
Congenital absence of vas deferens	CFTR	3909_3914delCTTGGAinsTGT | 3909_3914del6ins3 | 3909_3914delins	N/A	7	117292931	117292936
Cystic fibrosis	CFTR	3915TtoA | D1305E	N/A	7	117292937	117292937
Cystic fibrosis	CFTR	3917_3918dupCC | 3917_3918dup2 | 3917_3918dup	N/A	7	117292940	117292941
Congenital absence of vas deferens	CFTR	3920AtoG | Y1307C	N/A	7	117292942	117292942
Cystic fibrosis	CFTR	3921TtoA | Y1307X	N/A	7	117292943	117292943
Cystic fibrosis	CFTR	3922GtoT | E1308X	N/A	7	117292944	117292944
Congenital absence of vas deferens	CFTR	3927GtoT | Q1309H	N/A	7	117292949	117292949
Cystic fibrosis	CFTR	3929GtoA | W1310X	N/A	7	117292951	117292951
Cystic fibrosis	CFTR	3937CtoA | Q1313K	rs121909026	7	117292959	117292959
Cystic fibrosis	CFTR	3937CtoT | Q1313X	rs121909026	7	117292959	117292959
Cystic fibrosis	CFTR	3947GtoA | W1316X	rs121909010	7	117292969	117292969
Cystic fibrosis	CFTR	3953TtoC | V1318A	N/A	7	117292975	117292975
Cystic fibrosis	CFTR	3961GtoC | E1321Q	N/A	7	117292983	117292983
Cystic fibrosis	CFTR	3963plus1GtoC	N/A	7	117292986	117292986
Cystic fibrosis	CFTR	3963plus2TtoA	N/A	7	117292987	117292987
Exudative vitreoretinopathy	TSPAN12	313TtoC | C105R	N/A	7	120455830	120455830
Diabetes, type 1, association with	PAX4	962CtoA | P321H	rs712701	7	127251188	127251188
Retinitis pigmentosa	IMPDH1	602CtoT | T201M	N/A	7	128040571	128040571
Joubert syndrome	CEP41	536GtoA | R179H	N/A	7	130042527	130042527
Joubert syndrome	CEP41	423minus2AtoC	N/A	7	130042642	130042642
Distal renal tubular acidosis, autosomal recess	ATP6V0A4	1072CtoT | Q358X	N/A	7	138434020	138434020
Distal renal tubular acidosis, autosomal recess	ATP6V0A4	1030minus2AtoC	N/A	7	138434064	138434064
Distal renal tubular acidosis, autosomal recess	ATP6V0A4	828_831delAACA | 828_831del4 | 828_831del	N/A	7	138437568	138437571
Myotonia congenita	CLCN1	180plus3AtoT	N/A	7	143013488	143013488
Myotonia congenita	CLCN1	1111delC | 1111del1 | 1111del	N/A	7	143028690	143028690
Myotonia congenita	CLCN1	1117AtoT | M373L	N/A	7	143028696	143028696
Myotonia congenita	CLCN1	1129CtoT | R377X	N/A	7	143028708	143028708
Myotonia congenita	CLCN1	2124CtoG | F708L	N/A	7	143042807	143042807
Myotonia congenita	CLCN1	2149GtoT | E717X	N/A	7	143042832	143042832
Autism spectrum disorder, modifier of	CNTNAP2	824AtoG | H275R	N/A	7	146818140	146818140
Long QT syndrome	KCNH2	3196CtoG | L1066V	N/A	7	150644099	150644099
Long QT syndrome	KCNH2	3172dupG | 3172dup1 | 3172dup	N/A	7	150644122	150644123
Long QT syndrome	KCNH2	3167dupT | 3167dup1 | 3167dup	N/A	7	150644127	150644128
Long QT syndrome	KCNH2	865GtoA | E289K	N/A	7	150655198	150655198
Sudden infant death syndrome	KCNH2	835GtoA | V279M	N/A	7	150655228	150655228
Long QT syndrome	KCNH2	830CtoA | A277D	N/A	7	150655233	150655233
Long QT syndrome	KCNH2	826delT | 826del1 | 826del	N/A	7	150655237	150655237
Long QT syndrome	KCNH2	305_307dupATG | 305_307dup3 | 305_307dup	N/A	7	150671798	150671799
Long QT syndrome	KCNH2	305AtoC | D102A	N/A	7	150671801	150671801
Long QT syndrome	KCNH2	305AtoT | D102V	N/A	7	150671801	150671801
Long QT syndrome	KCNH2	301AtoG | K101E	N/A	7	150671805	150671805
Long QT syndrome	KCNH2	299GtoA | R100Q	N/A	7	150671807	150671807
Long QT syndrome	KCNH2	298CtoG | R100G	N/A	7	150671808	150671808
Long QT syndrome	KCNH2	298CtoT | R100W	N/A	7	150671808	150671808
Long QT syndrome	KCNH2	296AtoC | Y99S	N/A	7	150671810	150671810
Long QT syndrome	KCNH2	287TtoC | I96T	N/A	7	150671819	150671819
Long QT syndrome	KCNH2	286AtoG | I96V	N/A	7	150671820	150671820
Glaucoma, primary open angle	ASB10	1318AtoG | S440G	rs104886487	7	150873285	150873285
Holoprosencephaly	SHH	995TtoC | V332A	rs104894052	7	155595988	155595988
Holoprosencephaly	SHH	961CtoA | R321S	N/A	7	155596022	155596022
Holoprosencephaly	SHH	928CtoT | R310C	N/A	7	155596055	155596055
Holoprosencephaly	SHH	887GtoC | G296A	N/A	7	155596096	155596096
Currarino syndrome	MNX1	204_216dupGCCCGCCGACCGC | 204_216dup13 | 204_216dup	N/A	7	156802828	156802829
Currarino syndrome	MNX1	199_215dupGCTGCGCCCGCCGACCG | 199_215dup17 | 199_215dup	N/A	7	156802829	156802830
Currarino syndrome	MNX1	186dupC | 186dup1 | 186dup	N/A	7	156802858	156802859
Atrial fibrillation	GATA4	82CtoG | H28D	N/A	8	11565903	11565903
Congenital heart defects	GATA4	82CtoT | H28Y	N/A	8	11565903	11565903
Atrial fibrillation	GATA4	112TtoG | Y38D	N/A	8	11565933	11565933
Ventricular septal defect	GATA4	127CtoT | R43W	N/A	8	11565948	11565948
Congenital heart defects	GATA4	139_141delTCC | 139_141del3 | 139_141del	N/A	8	11565960	11565962

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Congenital heart defects	GATA4	155CtoT | S52F	rs104894074	8	11565976	11565976
Ventricular septal defect	GATA4	164AtoG | Q55R	N/A	8	11565985	11565985
Ventricular septal defect	GATA4	191GtoA | G64E	N/A	8	11566012	11566012
Congenital heart defects	GATA4	206GtoA | G69D	N/A	8	11566027	11566027
Atrial fibrillation	GATA4	209GtoC | S70T	N/A	8	11566030	11566030
Atrial septal defect	GATA4	259CtoT | P87S	N/A	8	11566080	11566080
Congenital heart defects	GATA4	270CtoA | S90R	N/A	8	11566091	11566091
Congenital heart defects	GATA4	278GtoC | G93A	rs56191129	8	11566099	11566099
Congenital heart defects	GATA4	284AtoG | D95G	N/A	8	11566105	11566105
Atrial fibrillation	GATA4	307CtoG | P103A	N/A	8	11566128	11566128
Atrial septal defect	GATA4	341_342insA | 341_342ins1 | 341_342ins	N/A	8	11566162	11566163
Congenital heart defects	GATA4	366_368dupCGC | 366_368dup3 | 366_368dup	N/A	8	11566189	11566190
Congenital heart defects	GATA4	369_374dupTGCCGC | 369_374dup6 | 369_374dup	N/A	8	11566195	11566196
Atrial fibrillation	GATA4	479GtoC | S160T	N/A	8	11566300	11566300
Congenital heart defects	GATA4	487CtoT | P163S	N/A	8	11566308	11566308
Congenital heart defects	GATA4	488CtoG | P163R	N/A	8	11566309	11566309
Atrial septal defect	GATA4	1060AtoG | T354A	N/A	8	11614506	11614506
Congenital heart defects	GATA4	1074delC | 1074del1 | 1074del	N/A	8	11614520	11614520
Congenital heart defects	GATA4	1075GtoA | E359K	N/A	8	11614521	11614521
Congenital heart defects	GATA4	1075delG | 1075del1 | 1075del	N/A	8	11614521	11614521
Multiple colorectal adenoma	NEIL2	770GtoT | R257L	rs8191664	8	11643553	11643553
Farber disease	ASAH1	67CtoG | H23D	N/A	8	17941501	17941501
Farber disease	ASAH1	66GtoC | Q22H	N/A	8	17941502	17941502
Lipoprotein lipase deficiency	LPL	42GtoA | W14X	N/A	8	19796993	19796993
Hypertriglyceridaemia	LPL	46_47delCA | 46_47del2 | 46_47del	N/A	8	19796997	19796998
Atrichia with papular lesions	HR	3470CtoG | P1157R	N/A	8	21973850	21973850
Atrichia with papular lesions	HR	3435delC | 3435del1 | 3435del	N/A	8	21973885	21973885
Parkinson disease	NEFM	2487_2489delTGT | 2487_2489del3 | 2487_2489del	N/A	8	24775855	24775857
Schizophrenia, association with	NRG1	881TtoC | M294T	rs10503929	8	32613983	32613983
Kallmann syndrome	FGFR1	2107GtoA | G703S	N/A	8	38271749	38271749
Kallmann syndrome	FGFR1	2107GtoC | G703R	N/A	8	38271749	38271749
Hypogonadotropic hypogonadism, idiopathic	FGFR1	2099CtoT | P700L	N/A	8	38271757	38271757
Kallmann syndrome	FGFR1	2077AtoT | I693F	N/A	8	38271779	38271779
Kallmann syndrome	FGFR1	2075AtoG | E692G	N/A	8	38271781	38271781
GnRH deficiency	FGFR1	165_171delTCGGCTG | 165_171del7 | 165_171del	N/A	8	38287387	38287393
Spherocytosis	ANK1	561_570delCACGGCTGCG | 561_570del10 | 561_570del	N/A	8	41583321	41583330
Spherocytosis	ANK1	520_539delCTCCCGGCCCTGCACATCGC | 520_539del20 | 520_539del	N/A	8	41583352	41583371
Dystonia, early-onset	THAP1	339TtoC | D113D	N/A	8	42693408	42693408
Mucopolysaccharidosis IIIC	HGSNAT	118plus1GtoA	N/A	8	42995758	42995758
Mucopolysaccharidosis IIIC	HGSNAT	1411GtoA | E471K	N/A	8	43048933	43048933
Mucopolysaccharidosis IIIC	HGSNAT	1441GtoT | V481L	N/A	8	43048963	43048963
Mucopolysaccharidosis IIIC	HGSNAT	1445TtoA | M482K	rs121908284	8	43048967	43048967
Mucopolysaccharidosis IIIC	HGSNAT	1457GtoA | G486E	N/A	8	43048979	43048979
Mucopolysaccharidosis IIIC	HGSNAT	1464plus1GtoA	N/A	8	43048987	43048987
Congenital anomalies of the kidney and urinar	SOX17	775TtoA | Y259N	N/A	8	55372085	55372085
CHARGE syndrome	CHD7	1714CtoT | Q572X	N/A	8	61693607	61693607
CHARGE syndrome	CHD7	1730dupA | 1730dup1 | 1730dup	N/A	8	61693623	61693624
CHARGE syndrome	CHD7	1735CtoT | Q579X	N/A	8	61693628	61693628
CHARGE syndrome	CHD7	1740delA | 1740del1 | 1740del	N/A	8	61693633	61693633
CHARGE syndrome	CHD7	1757_1770delACCTGCCATCAATA | 1757_1770del14 | 1757_1770del	N/A	8	61693650	61693663
CHARGE syndrome	CHD7	1953dupA | 1953dup1 | 1953dup	N/A	8	61693846	61693847
CHARGE syndrome	CHD7	1972GtoT | E658X	N/A	8	61693865	61693865
CHARGE syndrome	CHD7	1973_1974insT | 1973_1974ins1 | 1973_1974ins	N/A	8	61693866	61693867
CHARGE syndrome	CHD7	2034delA | 2034del1 | 2034del	N/A	8	61693927	61693927
CHARGE syndrome	CHD7	2095AtoG | S699G	N/A	8	61693988	61693988
CHARGE syndrome	CHD7	2096GtoA | S699N	N/A	8	61693989	61693989
CHARGE syndrome	CHD7	2096plus2TtoA	N/A	8	61693991	61693991
CHARGE syndrome	CHD7	2096plus2TtoC	N/A	8	61693991	61693991
CHARGE syndrome	CHD7	2374CtoT | Q792X	N/A	8	61713082	61713082
CHARGE syndrome	CHD7	6746delA | 6746del1 | 6746del	N/A	8	61766030	61766030
CHARGE syndrome	CHD7	6757GtoT | E2253X	N/A	8	61766041	61766041
CHARGE syndrome	CHD7	7165minus4AtoG	N/A	8	61769000	61769000
Spastic paraplegia	CYP7B1	889AtoG | T297A	N/A	8	65527751	65527751
Peroxisome biogenesis disorder	PEX2	739TtoC | C247R	rs61752128	8	77895676	77895676
Achromatopsia	CNGB3	29dupA | 29dup1 | 29dup	N/A	8	87755826	87755827
Ocular & skeletal abnormalities	GDF6	758AtoT | Q253L	rs121909355	8	97157401	97157401
Ocular & skeletal abnormalities	GDF6	647AtoG | D216G	N/A	8	97157512	97157512
Barrett oesophagus/oesophageal adenocarcin	CTHRC1	131AtoC | Q44P	N/A	8	104384015	104384015
Hypothyroidism	TG	5690GtoA | C1897Y	rs121912649	8	133980042	133980042
Goitre with hypothyroidism	TG	5747AtoG | Y1916C	N/A	8	133980099	133980099
Hypothyroidism	TG	5791AtoG | I1931V	rs115877910	8	133980143	133980143
Chylomicronaemia	GPIHBP1	194GtoA | C65Y	N/A	8	144296900	144296900
Chylomicronaemia	GPIHBP1	194GtoC | C65S	N/A	8	144296900	144296900
Chylomicronaemia	GPIHBP1	202TtoG | C68G	N/A	8	144296908	144296908
Chylomicronaemia	GPIHBP1	203GtoA | C68Y	N/A	8	144296909	144296909
Leukemia, risk, association with	TOP1MT	1573CtoT | R525W	rs2293925	8	144392368	144392368
Epidermolysis bullosa simplex	PLEC	2681_2693+1delAGGCCGTCACCAGG | 2681_2693+1del14 | 2681_2693+1del	N/A	8	145004311	145004324
Muscular dystrophy with epidermolysis bullos	PLEC	2677_2685delCAGGAGGCC | 2677_2685del9 | 2677_2685del	N/A	8	145004320	145004328
Congenital heart defects	FOXH1	584GtoT | G195V	N/A	8	145700135	145700135
Nicholaides-Baraitser syndrome	SMARCA2	3436AtoC | S1146R	N/A	9	2110397	2110397
Nicholaides-Baraitser syndrome	SMARCA2	3562GtoC | A1188P	N/A	9	2115927	2115927
Nicholaides-Baraitser syndrome	SMARCA2	3563CtoA | A1188E	N/A	9	2115928	2115928
Nicholaides-Baraitser syndrome	SMARCA2	3602CtoT | A1201V	N/A	9	2115967	2115967
Nicholaides-Baraitser syndrome	SMARCA2	3604GtoT | G1202C	N/A	9	2115969	2115969
Hyperglycinaemia, non-ketotic	GLDC	1175delC | 1175del1 | 1175del	N/A	9	6595100	6595100
Hyperglycinaemia, non-ketotic	GLDC	1166CtoT | A389V	rs121964979	9	6595109	6595109
Hyperglycinaemia, non-ketotic	GLDC	937GtoC | A313P	N/A	9	6604709	6604709
Hyperglycinaemia, non-ketotic	GLDC	28delC | 28del1 | 28del	N/A	9	6645472	6645472
Friedreich ataxia	FXN	100delG | 100del1 | 100del	rs138837292	9	71650798	71650798
Friedreich ataxia	FXN	104delC | 104del1 | 104del	rs140510894	9	71650802	71650802
Hepatitis B, chronic, association with	TMEM2	3761GtoA | S1254N	N/A	9	74305098	74305098
Deafness	TMC1	100CtoT | R34X	rs121908073	9	75309494	75309494
Hypomagnesaemia with secondary hypocalcae	TRPM6	2667plus1GtoA	N/A	9	77403529	77403529
Holoprosencephaly	GAS1	133CtoG | R45G	N/A	9	89561562	89561562
Fructose-1,6-bisphosphatase deficiency	FBP1	685CtoT | Q229X	N/A	9	97369117	97369117

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Fructose-1,6-bisphosphatase deficiency	FBP1	648CtoG | Y216X	N/A	9	97369154	97369154
Fructose-1,6-bisphosphatase deficiency	FBP1	639CtoG | N213K	N/A	9	97369163	97369163
Fructose-1,6-bisphosphatase deficiency	FBP1	616_619delAAAG | 616_619del4 | 616_619del	N/A	9	97369183	97369186
Nevoid basal cell carcinoma syndrome	PTCH1	1501_1503+5delCAGGTACT | 1501_1503+5del8 | 1501_1503+5del	N/A	9	98239824	98239831
Nevoid basal cell carcinoma syndrome	PTCH1	1503plus1GtoC	N/A	9	98239828	98239828
Nevoid basal cell carcinoma syndrome	PTCH1	1503GtoA	N/A	9	98239829	98239829
Nevoid basal cell carcinoma syndrome	PTCH1	1472delT | 1472del1 | 1472del	N/A	9	98239860	98239860
Nevoid basal cell carcinoma syndrome	PTCH1	1470_1471delGAinsTT | 1470_1471del2ins2 | 1470_1471delins	N/A	9	98239861	98239862
Nevoid basal cell carcinoma syndrome	PTCH1	1467dupA | 1467dup1 | 1467dup	N/A	9	98239864	98239865
Nevoid basal cell carcinoma syndrome	PTCH1	1462_1463dupTG | 1462_1463dup2 | 1462_1463dup	N/A	9	98239868	98239869
Nevoid basal cell carcinoma syndrome	PTCH1	1451GtoT | G484V	N/A	9	98239881	98239881
Nevoid basal cell carcinoma syndrome	PTCH1	1450GtoA | G484R	N/A	9	98239882	98239882
Nevoid basal cell carcinoma syndrome	PTCH1	1436TtoG | L479R	N/A	9	98239896	98239896
Nevoid basal cell carcinoma syndrome	PTCH1	1416_1419dupTGGC | 1416_1419dup4 | 1416_1419dup	N/A	9	98239912	98239913
Nevoid basal cell carcinoma syndrome	PTCH1	1416_1419delTGGC | 1416_1419del4 | 1416_1419del	N/A	9	98239913	98239916
Nevoid basal cell carcinoma syndrome	PTCH1	1415CtoA | A472D	N/A	9	98239917	98239917
Nevoid basal cell carcinoma syndrome	PTCH1	1414delG | 1414del1 | 1414del	N/A	9	98239918	98239918
Nevoid basal cell carcinoma syndrome	PTCH1	1380GtoA | W460X	N/A	9	98239952	98239952
Nevoid basal cell carcinoma syndrome	PTCH1	1340_1343delTACT | 1340_1343del4 | 1340_1343del	N/A	9	98240341	98240344
Gorlin-syndrome-related odontogenic keratoc	PTCH1	1338_1339insGCG | 1338_1339ins3 | 1338_1339ins	N/A	9	98240345	98240346
Holoprosencephaly	PTCH1	1328CtoG | A443G	N/A	9	98240356	98240356
Nevoid basal cell carcinoma syndrome	PTCH1	1325TtoA | V442E	N/A	9	98240359	98240359
Nevoid basal cell carcinoma syndrome	PTCH1	1291delC | 1291del1 | 1291del	N/A	9	98240393	98240393
Nevoid basal cell carcinoma syndrome	PTCH1	1261dupT | 1261dup1 | 1261dup	N/A	9	98240422	98240423
Nevoid basal cell carcinoma syndrome	PTCH1	331delG | 331del1 | 331del	N/A	9	98268752	98268752
Nevoid basal cell carcinoma syndrome	PTCH1	328GtoC | G110R	N/A	9	98268755	98268755
Nevoid basal cell carcinoma syndrome	PTCH1	323_324insGA | 323_324ins2 | 323_324ins	N/A	9	98268759	98268760
Nevoid basal cell carcinoma syndrome	PTCH1	317TtoG | L106R	N/A	9	98268766	98268766
Xeroderma pigmentosum (A)	XPA	470_489delAAAGAGAGCCACCTCTTAAA | 470_489del20 | 470_489del	N/A	9	100449444	100449463
Nephronophthisis 2	INVS	1948GtoC | A650P	rs147452898	9	103046765	103046765
Fructose intolerance	ALDOB	911GtoA | R304Q	rs145078268	9	104187213	104187213
Fructose intolerance	ALDOB	910CtoT | R304W	N/A	9	104187214	104187214
Reduced plasma HDL cholesterol	ABCA1	2789CtoT | S930F	N/A	9	107584816	107584816
Tangier disease	ABCA1	2786CtoT | T929I	N/A	9	107584819	107584819
Increased plasma HDL cholesterol	ABCA1	2660GtoT | C887F	N/A	9	107584945	107584945
Primary amenorrhea	NR5A1	1138GtoT | D380Y	N/A	9	127253360	127253360
Primary amenorrhea	NR5A1	1052CtoT | A351V	N/A	9	127253446	127253446
Nail patella syndrome	LMX1B	622_624delGGCinsA | 622_624del3ins1 | 622_624delins	N/A	9	129455483	129455485
Nail patella syndrome	LMX1B	641delA | 641del1 | 641del	N/A	9	129455502	129455502
Nail patella syndrome	LMX1B	661CtoT | R221X	rs121909487	9	129455522	129455522
Nail patella syndrome	LMX1B	661delC | 661del1 | 661del	N/A	9	129455522	129455522
Nail patella syndrome	LMX1B	663dupA | 663dup1 | 663dup	N/A	9	129455524	129455525
Nail patella syndrome	LMX1B	667CtoG | R223G	N/A	9	129455528	129455528
Nail patella syndrome	LMX1B	667CtoT | R223W	N/A	9	129455528	129455528
Nail patella syndrome	LMX1B	668GtoA | R223Q	rs121909491	9	129455529	129455529
Nail patella syndrome	LMX1B	672dupC | 672dup1 | 672dup	N/A	9	129455533	129455534
Nail patella syndrome	LMX1B	676_681delCTCACC | 676_681del6 | 676_681del	N/A	9	129455537	129455542
Nail patella syndrome	LMX1B	680_686delCCACGCA | 680_686del7 | 680_686del	N/A	9	129455541	129455547
Nail patella syndrome	LMX1B	691CtoT | R231X	rs121909490	9	129455552	129455552
Nail patella syndrome	LMX1B	692GtoC | R231P	N/A	9	129455553	129455553
Nail patella syndrome	LMX1B	706GtoC | A236P	N/A	9	129455567	129455567
Nail patella syndrome	LMX1B	715GtoT | E239X	N/A	9	129455576	129455576
Nail patella syndrome	LMX1B	720_722delCTC | 720_722del3 | 720_722del	N/A	9	129455581	129455583
Nail patella syndrome	LMX1B	721TtoC | S241P	N/A	9	129455582	129455582
Nail patella syndrome	LMX1B	722CtoA | S241X	N/A	9	129455583	129455583
Nail patella syndrome	LMX1B	725CtoA | S242X	N/A	9	129455586	129455586
Nail patella syndrome	LMX1B	728delA | 728del1 | 728del	N/A	9	129455589	129455589
Mental retardation & nonsyndromic epilepsy	STXBP1	1162CtoT | R388X	rs121918321	9	130438134	130438134
Intellectual disability, nonsyndromic without e	STXBP1	1206delT | 1206del1 | 1206del	N/A	9	130438178	130438178
Epileptic encephalopathy, early infantile	STXBP1	1217GtoA | R406H	N/A	9	130438189	130438189
Haemorrhagic telangiectasia 1	ENG	1311plus2TtoA	rs111694746	9	130581899	130581899
Haemorrhagic telangiectasia 1	ENG	1311plus2TtoC	N/A	9	130581899	130581899
Haemorrhagic telangiectasia	ENG	1311plus2TtoG	N/A	9	130581899	130581899
Haemorrhagic telangiectasia 1	ENG	1311+1dupG | 1311+1dup1 | 1311+1dup	N/A	9	130581899	130581900
Haemorrhagic telangiectasia 1	ENG	1311+1delG | 1311+1del1 | 1311+1del	N/A	9	130581900	130581900
Haemorrhagic telangiectasia 1	ENG	1311GtoA	N/A	9	130581901	130581901
Haemorrhagic telangiectasia 1	ENG	1311GtoC	N/A	9	130581901	130581901
Haemorrhagic telangiectasia	ENG	1311GtoT	N/A	9	130581901	130581901
Haemorrhagic telangiectasia	ENG	1310GtoA | R437Q	N/A	9	130581902	130581902
Haemorrhagic telangiectasia 1	ENG	1309CtoT | R437W	N/A	9	130581903	130581903
Haemorrhagic telangiectasia 1	ENG	1306CtoT | Q436X	N/A	9	130581906	130581906
Haemorrhagic telangiectasia 1	ENG	1292CtoA | S431X	N/A	9	130581920	130581920
Haemorrhagic telangiectasia 1	ENG	1274CtoG | A425G	N/A	9	130581938	130581938
Haemorrhagic telangiectasia 1	ENG	1273minus1GtoA	N/A	9	130581940	130581940
Haemorrhagic telangiectasia 1	ENG	1273minus1GtoC	N/A	9	130581940	130581940
Haemorrhagic telangiectasia 1	ENG	1273minus2AtoG	N/A	9	130581941	130581941
Haemorrhagic telangiectasia 1	ENG	736delG | 736del1 | 736del	N/A	9	130587590	130587590
Haemorrhagic telangiectasia 1	ENG	360+4_360+7delAGTG | 360+4_360+7del4 | 360+4_360+7del	N/A	9	130591959	130591962
Haemorrhagic telangiectasia 1	ENG	360plus5GtoA	N/A	9	130591961	130591961
Haemorrhagic telangiectasia 1	ENG	360plus4AtoG	N/A	9	130591962	130591962
Haemorrhagic telangiectasia 1	ENG	360plus1GtoA	N/A	9	130591965	130591965
Haemorrhagic telangiectasia 1	ENG	360plus1GtoC	N/A	9	130591965	130591965
Haemorrhagic telangiectasia 1	ENG	360plus1GtoT	N/A	9	130591965	130591965
Haemorrhagic telangiectasia 1	ENG	360CtoA | Y120X	rs121918402	9	130591966	130591966
Haemorrhagic telangiectasia 1	ENG	344delC | 344del1 | 344del	N/A	9	130591982	130591982
Haemorrhagic telangiectasia 1	ENG	332_338delCCCTGGG | 332_338del7 | 332_338del	N/A	9	130591988	130591994
Haemorrhagic telangiectasia 1	ENG	338delG | 338del1 | 338del	N/A	9	130591988	130591988
Haemorrhagic telangiectasia	ENG	331GtoA | A111T	N/A	9	130591995	130591995
Haemorrhagic telangiectasia 1	ENG	326_327delTC | 326_327del2 | 326_327del	N/A	9	130591999	130592000
Haemorrhagic telangiectasia 1	ENG	324dupT | 324dup1 | 324dup	N/A	9	130592001	130592002
Haemorrhagic telangiectasia 1	ENG	320TtoG | L107R	N/A	9	130592006	130592006
Haemorrhagic telangiectasia 1	ENG	314TtoA | V105D	N/A	9	130592012	130592012
Haemorrhagic telangiectasia 1	ENG	309_311delCAG | 309_311del3 | 309_311del	N/A	9	130592015	130592017
Haemorrhagic telangiectasia	ENG	290TtoG | L97R	N/A	9	130592036	130592036
Haemorrhagic telangiectasia 1	ENG	287TtoC | L96P	N/A	9	130592039	130592039

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Haemorrhagic telangiectasia 1	ENG	260_277dupAAAATGGCACCTGGCCCC | 260_277dup18 | 260_277dup	N/A	9	130592048	130592049
Haemorrhagic telangiectasia 1	ENG	277CtoT | R93X	N/A	9	130592049	130592049
Haemorrhagic telangiectasia 1	ENG	277delC | 277del1 | 277del	N/A	9	130592049	130592049
Haemorrhagic telangiectasia 1	ENG	166CtoT | Q56X	N/A	9	130605426	130605426
Haemorrhagic telangiectasia	ENG	164delC | 164del1 | 164del	N/A	9	130605428	130605428
Haemorrhagic telangiectasia 1	ENG	159CtoA | C53X	N/A	9	130605433	130605433
Haemorrhagic telangiectasia 1	ENG	157TtoC | C53R	N/A	9	130605435	130605435
Haemorrhagic telangiectasia 1	ENG	155GtoA | G52D	N/A	9	130605437	130605437
Haemorrhagic telangiectasia 1	ENG	155GtoT | G52V	N/A	9	130605437	130605437
Haemorrhagic telangiectasia 1	ENG	154GtoC | G52R	N/A	9	130605438	130605438
Ichthyosis prematurity syndrome	SLC27A4	103GtoT | G35X	N/A	9	131105514	131105514
Ichthyosis prematurity syndrome	SLC27A4	1430TtoA | V477D	rs147688821	9	131117437	131117437
Citrullinaemia	ASS1	323GtoT | R108L	rs35269064	9	133333936	133333936
Citrullinaemia	ASS1	349GtoA | G117S	N/A	9	133333962	133333962
Citrullinaemia	ASS1	350GtoA | G117D	N/A	9	133333963	133333963
Citrullinaemia	ASS1	352GtoA | A118T	N/A	9	133333965	133333965
Citrullinaemia	ASS1	356CtoT | T119I	N/A	9	133333969	133333969
Bilateral occipital pachygyria	LAMC3	903_904delTG | 903_904del2 | 903_904del	N/A	9	133911646	133911647
Motor disability, microcephaly, mental retarda	POMT1	1241plus1GtoA	N/A	9	134388719	134388719
Tuberous sclerosis	TSC1	2145delG | 2145del1 | 2145del	rs118203650	9	135779101	135779101
Tuberous sclerosis	TSC1	2135_2139delATGCC | 2135_2139del5 | 2135_2139del	rs118203649	9	135779107	135779111
Tuberous sclerosis	TSC1	2131CtoT | Q711X	rs118203647	9	135779115	135779115
Tuberous sclerosis	TSC1	2112TtoA | Y704X	rs118203646	9	135779134	135779134
Tuberous sclerosis	TSC1	2111_2112delAT | 2111_2112del2 | 2111_2112del	rs118203645	9	135779134	135779135
Tuberous sclerosis	TSC1	2109_2110delCT | 2109_2110del2 | 2109_2110del	rs118203643	9	135779136	135779137
Tuberous sclerosis	TSC1	2103_2109dupGTTACTC | 2103_2109dup7 | 2103_2109dup	N/A	9	135779136	135779137
Tuberous sclerosis	TSC1	2103_2106dupGTTA | 2103_2106dup4 | 2103_2106dup	N/A	9	135779139	135779140
Tuberous sclerosis	TSC1	2106_2107insGTTTA | 2106_2107ins5 | 2106_2107ins	N/A	9	135779139	135779140
Tuberous sclerosis	TSC1	2103_2106delGTTA | 2103_2106del4 | 2103_2106del	rs118203640	9	135779140	135779143
Tuberous sclerosis	TSC1	2106delAinsGT | 2106del1ins2 | 2106delins	N/A	9	135779140	135779140
Tuberous sclerosis	TSC1	1533delT | 1533del1 | 1533del	rs118203545	9	135781432	135781432
Tuberous sclerosis	TSC1	1530_1531delCA | 1530_1531del2 | 1530_1531del	N/A	9	135781434	135781435
Tuberous sclerosis	TSC1	1525CtoT | R509X	rs118203542	9	135781440	135781440
Tuberous sclerosis	TSC1	1515_1516dupTC | 1515_1516dup2 | 1515_1516dup	N/A	9	135781448	135781449
Tuberous sclerosis	TSC1	1515dupT | 1515dup1 | 1515dup	N/A	9	135781449	135781450
Tuberous sclerosis	TSC1	1503delA | 1503del1 | 1503del	N/A	9	135781462	135781462
Tuberous sclerosis	TSC1	1501GtoT | G501X	N/A	9	135781464	135781464
Tuberous sclerosis	TSC1	1499GtoA | R500Q	rs118203538	9	135781466	135781466
Tuberous sclerosis	TSC1	1498CtoT | R500X	rs118203537	9	135781467	135781467
Tuberous sclerosis	TSC1	1490_1491delTG | 1490_1491del2 | 1490_1491del	rs118203536	9	135781474	135781475
Tuberous sclerosis	TSC1	912_913+5delTGGTAAA | 912_913+5del7 | 912_913+5del	N/A	9	135787664	135787670
Tuberous sclerosis	TSC1	901_913+1delCAGAATAGCTATGG | 901_913+1del14 | 901_913+1del	N/A	9	135787668	135787681
Tuberous sclerosis	TSC1	913GtoA | G305R	N/A	9	135787669	135787669
Tuberous sclerosis	TSC1	913GtoT | G305W	N/A	9	135787669	135787669
Tuberous sclerosis	TSC1	913GtoA	rs118203468	9	135787669	135787669
Tuberous sclerosis	TSC1	913GtoT	rs118203468	9	135787669	135787669
Tuberous sclerosis	TSC1	901_902delCA | 901_902del2 | 901_902del	rs118203464	9	135787680	135787681
Tuberous sclerosis	TSC1	901CtoT | Q301X	rs118203463	9	135787681	135787681
Leigh syndrome	SURF1	55minus1GtoA	N/A	9	136223176	136223176
Leigh syndrome	SURF1	19_35dupTTGCAGCTGGGGCTGCG | 19_35dup17 | 19_35dup	N/A	9	136223294	136223295
Leigh syndrome	SURF1	23AtoT | Q8L	N/A	9	136223307	136223307
Thrombotic thrombocytopaenic purpura	ADAMTS13	237CtoG | I79M	N/A	9	136289505	136289505
Thrombotic thrombocytopaenic purpura	ADAMTS13	687minus2AtoG	N/A	9	136293752	136293752
Thrombotic thrombocytopaenic purpura	ADAMTS13	695TtoA | L232Q	N/A	9	136293762	136293762
Upshaw-Schulman syndrome	ADAMTS13	701AtoG | H234R	N/A	9	136293768	136293768
Upshaw-Schulman syndrome	ADAMTS13	702CtoA | H234Q	N/A	9	136293769	136293769
Thrombotic thrombocytopaenic purpura	ADAMTS13	703GtoC | D235H	N/A	9	136293770	136293770
Thrombotic thrombocytopaenic purpura	ADAMTS13	703GtoT | D235Y	N/A	9	136293770	136293770
Upshaw-Schulman syndrome	ADAMTS13	706GtoT | G236C	N/A	9	136293773	136293773
Thrombotic thrombocytopaenic purpura	ADAMTS13	724_729delTGCGGC | 724_729del6 | 724_729del	N/A	9	136293791	136293796
Thrombotic thrombocytopaenic purpura	ADAMTS13	749CtoT | A250V	rs121908478	9	136293816	136293816
Thrombotic thrombocytopaenic purpura	ADAMTS13	768_774delCCGCGCC | 768_774del7 | 768_774del	N/A	9	136293835	136293841
Thrombotic thrombocytopaenic purpura	ADAMTS13	788CtoG | S263C	N/A	9	136293855	136293855
Thrombotic thrombocytopaenic purpura	ADAMTS13	803GtoC | R268P	rs121908477	9	136293870	136293870
Thrombotic thrombocytopaenic purpura	ADAMTS13	1039TtoA | C347S	N/A	9	136297760	136297760
Thrombotic thrombocytopaenic purpura	ADAMTS13	1520GtoA | R507Q	N/A	9	136302953	136302953
Thrombotic thrombocytopaenic purpura	ADAMTS13	1523GtoA | C508Y	N/A	9	136302956	136302956
Reduced activity	ADAMTS13	1852CtoG | P618A	rs28647808	9	136305530	136305530
Upshaw-Schulman syndrome	ADAMTS13	1892CtoT | A631V	N/A	9	136305570	136305570
Thrombotic thrombocytopaenic purpura	ADAMTS13	2195CtoT | A732V	rs41314453	9	136307825	136307825
Thrombotic thrombocytopaenic purpura	ADAMTS13	3716GtoT | G1239V	N/A	9	136321665	136321665
Thrombotic thrombocytopaenic purpura	ADAMTS13	3735GtoA | W1245X	N/A	9	136321684	136321684
Thrombotic thrombocytopaenic purpura	ADAMTS13	4143dupA | 4143dup1 | 4143dup	N/A	9	136324161	136324162
Ehlers-Danlos syndrome	COL5A1	3184CtoT | R1062X	N/A	9	137696890	137696890
Ehlers-Danlos syndrome	COL5A1	3333_3342delACCCCCAGGG | 3333_3342del10 | 3333_3342del	N/A	9	137698109	137698118
Pituitary hormone deficiency, combined	LHX3	368GtoA | C123Y	N/A	9	139091625	139091625
Pituitary hormone deficiency, combined	LHX3	347AtoG | Y116C	N/A	9	139091646	139091646
Pituitary hormone deficiency, combined	LHX3	302_303delGCinsTCCT | 302_303del2ins4 | 302_303delins	N/A	9	139091690	139091691
Joubert syndrome	INPP5E	1304GtoA | R435Q	rs121918129	9	139327014	139327014
Berardinelli-Seip lipodystrophy	AGPAT2	755_763delTGAGGACCA | 755_763del9 | 755_763del	N/A	9	139568278	139568286
Berardinelli-Seip lipodystrophy	AGPAT2	716CtoT | A239V	rs145975461	9	139568325	139568325
Berardinelli-Seip lipodystrophy	AGPAT2	713CtoG | A238G	N/A	9	139568328	139568328
Berardinelli-Seip lipodystrophy	AGPAT2	683TtoC | L228P	rs104894100	9	139568358	139568358
Berardinelli-Seip lipodystrophy	AGPAT2	646AtoT | K216X	rs138994150	9	139569202	139569202
Berardinelli-Seip lipodystrophy	AGPAT2	589minus2AtoG	rs116807569	9	139569261	139569261
Berardinelli-Seip lipodystrophy	AGPAT2	570CtoA | Y190X	rs121908926	9	139571055	139571055
Berardinelli-Seip lipodystrophy	AGPAT2	538delG | 538del1 | 538del	N/A	9	139571087	139571087
Deafness, nonsyndromic	TPRN	227_237dupGGGCGCGGCTG | 227_237dup11 | 227_237dup	N/A	9	140094926	140094927
Deafness, nonsyndromic	TPRN	225_235delGGGGGCGCGGC | 225_235del11 | 225_235del	N/A	9	140094929	140094939
Hereditary hypophosphataemic rickets with hy	SLC34A3	908delC | 908del1 | 908del	N/A	9	140128376	140128376
Brugada syndrome	CACNB2	32CtoT | T11I	N/A	10	18629887	18629887
Pancreatic and cerebellar agenesis	PTF1A	705dupG | 705dup1 | 705dup	N/A	10	23482164	23482165
Cerebellar agenesis	PTF1A	720delG | 720del1 | 720del	N/A	10	23482179	23482179
Posterior polymorphous corneal dystrophy	ZEB1	1AtoG | M1V	N/A	10	31608164	31608164
Posterior polymorphous corneal dystrophy	ZEB1	2TtoG | M1R	N/A	10	31608165	31608165

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Posterior polymorphous corneal dystrophy	ZEB1	34CtoT | Q12X	N/A	10	31608197	31608197
Corneal dystrophy, Fuchs late-onset	ZEB1	233AtoG | N78S	N/A	10	31750140	31750140
Hirschsprung disease, association with	RET	NULL	N/A	10	43572512	43572512
Hirschsprung disease, association with	RET	NULL	N/A	10	43572516	43572516
Hirschsprung disease	RET	898GtoA | D300N	N/A	10	43601854	43601854
Hirschsprung disease	RET	905_906insGCAG | 905_906ins4 | 905_906ins	N/A	10	43601861	43601862
Hirschsprung disease	RET	937CtoT | R313W	N/A	10	43601893	43601893
Hirschsprung disease	RET	938GtoA | R313Q	rs77702891	10	43601894	43601894
Hirschsprung disease	RET	947GtoT | S316I	N/A	10	43601903	43601903
Thyroid carcinoma, medullary	RET	961GtoA | G321R	N/A	10	43601917	43601917
Hirschsprung disease	RET	972GtoC | W324C	N/A	10	43601928	43601928
Hirschsprung disease	RET	981GtoA	N/A	10	43601937	43601937
Hirschsprung disease	RET	988CtoT | R330W	N/A	10	43601944	43601944
Hirschsprung disease	RET	988dupC | 988dup1 | 988dup	N/A	10	43601944	43601945
Hirschsprung disease	RET	989GtoA | R330Q	rs80236571	10	43601945	43601945
Hirschsprung disease	RET	991GtoA | V331M	N/A	10	43601947	43601947
Hirschsprung disease	RET	1776_1777delGGinsAT | 1776_1777del2ins2 | 1776_1777delins	N/A	10	43609020	43609021
Hirschsprung disease	RET	1783GtoC	N/A	10	43609027	43609027
Thyroid cancer	RET	1807AtoC | K603Q	N/A	10	43609051	43609051
Hirschsprung disease	RET	1809delA | 1809del1 | 1809del	N/A	10	43609053	43609053
Thyroid carcinoma, medullary	RET	1817AtoG | Y606C	N/A	10	43609061	43609061
Hirschsprung disease	RET	1825TtoA | C609S	N/A	10	43609069	43609069
Multiple endocrine neoplasia 2A	RET	1825TtoC | C609R	rs77558292	10	43609069	43609069
Multiple endocrine neoplasia 2A	RET	1825TtoG | C609G	N/A	10	43609069	43609069
Thyroid cancer	RET	1826GtoA | C609Y	rs77939446	10	43609070	43609070
Thyroid cancer	RET	1826GtoC | C609S	N/A	10	43609070	43609070
Thyroid cancer	RET	1826GtoT | C609F	N/A	10	43609070	43609070
Hirschsprung disease	RET	1827CtoG | C609W	N/A	10	43609071	43609071
Multiple endocrine neoplasia 2A	RET	1831TtoA | C611S	N/A	10	43609075	43609075
Multiple endocrine neoplasia 2A	RET	1831TtoC | C611R	N/A	10	43609075	43609075
Thyroid cancer	RET	1831TtoG | C611G	N/A	10	43609075	43609075
Multiple endocrine neoplasia 2A	RET	1832GtoA | C611Y	N/A	10	43609076	43609076
Multiple endocrine neoplasia 2A	RET	1832GtoT | C611F	N/A	10	43609076	43609076
Thyroid carcinoma, medullary	RET	1832_1833delGCinsAT | 1832_1833del2ins2 | 1832_1833delins	N/A	10	43609076	43609077
Hirschsprung disease	RET	1832_1833delGCinsCT | 1832_1833del2ins2 | 1832_1833delins	N/A	10	43609076	43609077
Multiple endocrine neoplasia 2A	RET	1833CtoG | C611W	rs80069458	10	43609077	43609077
Hirschsprung disease	RET	1836_1842delCCCTGAG | 1836_1842del7 | 1836_1842del	N/A	10	43609080	43609086
Thyroid cancer	RET	1852TtoA | C618S	N/A	10	43609096	43609096
Thyroid cancer	RET	1852TtoC | C618R	rs76262710	10	43609096	43609096
Multiple endocrine neoplasia 2A	RET	1852TtoG | C618G	rs76262710	10	43609096	43609096
Thyroid carcinoma, medullary	RET	1853GtoA | C618Y	N/A	10	43609097	43609097
Multiple endocrine neoplasia 2A	RET	1853GtoC | C618S	rs79781594	10	43609097	43609097
Multiple endocrine neoplasia 2A	RET	1853GtoT | C618F	N/A	10	43609097	43609097
Multiple endocrine neoplasia 2	RET	1854CtoA | C618X	N/A	10	43609098	43609098
Thyroid cancer	RET	1858TtoA | C620S	N/A	10	43609102	43609102
Thyroid cancer	RET	1858TtoC | C620R	rs77316810	10	43609102	43609102
Multiple endocrine neoplasia 2A	RET	1858TtoG | C620G	N/A	10	43609102	43609102
Multiple endocrine neoplasia 2A	RET	1859GtoA | C620Y	rs77503355	10	43609103	43609103
Thyroid cancer	RET	1859GtoC | C620S	rs77503355	10	43609103	43609103
Thyroid cancer	RET	1859GtoT | C620F	rs77503355	10	43609103	43609103
Thyroid cancer	RET	1860CtoG | C620W	rs79890926	10	43609104	43609104
Hirschsprung disease	RET	1876CtoA | Q626K	N/A	10	43609120	43609120
Hirschsprung disease	RET	1879plus1GtoA	N/A	10	43609124	43609124
Hirschsprung disease	RET	1879plus2TtoA	N/A	10	43609125	43609125
Multifollicular thyroid carcinoma	RET	1879plus4AtoG	N/A	10	43609127	43609127
Hirschsprung disease	RET	1879plus5GtoC	N/A	10	43609128	43609128
Hirschsprung disease	RET	2975CtoT | P992L	N/A	10	43620366	43620366
Cockayne syndrome	ERCC6	2870TtoG | V957G	N/A	10	50680476	50680476
Cockayne syndrome	ERCC6	2867_2870delAAGT | 2867_2870del4 | 2867_2870del	N/A	10	50680476	50680479
Congenital myasthenic syndrome with episodi	CHAT	1007TtoC | I336T	rs121912823	10	50835727	50835727
Congenital myasthenic syndrome with episodi	CHAT	1061CtoT | T354M	N/A	10	50835781	50835781
Congenital myasthenic syndrome with episodi	CHAT	1321GtoA | E441K	rs121912816	10	50856592	50856592
Tetralogy of Fallot	NODAL	179GtoT | S60I	N/A	10	72201245	72201245
Reduced activity	NODAL	176GtoT | R59L	N/A	10	72201248	72201248
Tetralogy of Fallot	NODAL	164CtoT | A55V	N/A	10	72201260	72201260
Haemophagocytic lymphohistiocytosis, familia	PRF1	305GtoT | C102F	N/A	10	72360354	72360354
Haemophagocytic lymphohistiocytosis, familia	PRF1	283TtoC | W95R	N/A	10	72360376	72360376
Childhood acute lymphoblastic leukemia, asso	PRF1	272CtoT | A91V	rs35947132	10	72360387	72360387
Haemophagocytic lymphohistiocytosis, familia	PRF1	265CtoA | P89T	N/A	10	72360394	72360394
Haemophagocytic lymphohistiocytosis, familia	PRF1	217TtoC | C73R	N/A	10	72360442	72360442
Haemophagocytic lymphohistiocytosis, familia	PRF1	209AtoT | D70V	N/A	10	72360450	72360450
Haemophagocytic lymphohistiocytosis, familia	PRF1	208GtoT | D70Y	N/A	10	72360451	72360451
Haemophagocytic lymphohistiocytosis, familia	PRF1	207delC | 207del1 | 207del	N/A	10	72360452	72360452
Haemophagocytic lymphohistiocytosis, familia	PRF1	185_195delACACACAAAGG | 185_195del11 | 185_195del	N/A	10	72360464	72360474
Haemophagocytic lymphohistiocytosis, familia	PRF1	190CtoT | Q64X	rs104894180	10	72360469	72360469
Haemophagocytic lymphohistiocytosis, familia	PRF1	150delG | 150del1 | 150del	N/A	10	72360509	72360509
Haemophagocytic lymphohistiocytosis, familia	PRF1	148GtoA | V50M	N/A	10	72360511	72360511
Haemophagocytic lymphohistiocytosis, familia	PRF1	93CtoG | C31W	N/A	10	72360566	72360566
Haemophagocytic lymphohistiocytosis, familia	PRF1	83GtoA | R28H	N/A	10	72360576	72360576
Haemophagocytic lymphohistiocytosis, familia	PRF1	10CtoT | R4C	rs12161733	10	72360649	72360649
Haemophagocytic lymphohistiocytosis, familia	PRF1	3GtoA | M1I	N/A	10	72360656	72360656
Haemophagocytic lymphohistiocytosis, familia	PRF1	1AtoG | M1V	N/A	10	72360658	72360658
Hearing loss, non-syndromic	CDH23	2407GtoA | V803I	N/A	10	73461788	73461788
Metachromatic leukodystrophy	PSAP	1AtoG | M1V	N/A	10	73610978	73610978
Prosaposin deficiency	PSAP	1AtoT | M1L	rs121918106	10	73610978	73610978
Hypomyelination, hypodontia & hypogonadot	POLR3A	2324AtoT | N775I	N/A	10	79761990	79761990
Methionine adenosyltransferase deficiency	MAT1A	890CtoA | A297D	N/A	10	82034834	82034834
Homocystinuria	MAT1A	874CtoT | R292C	N/A	10	82034850	82034850
Methionine adenosyltransferase deficiency	MAT1A	867GtoT | K289N	N/A	10	82034857	82034857
Methionine adenosyltransferase deficiency	MAT1A	776CtoT | A259V	rs138556525	10	82034948	82034948
Methionine adenosyltransferase deficiency	MAT1A	773AtoG | D258G	N/A	10	82034951	82034951
Methionine adenosyltransferase deficiency	MAT1A	769GtoA | G257R	N/A	10	82034955	82034955
Brachyolmia	PAPSS2	381+2delT | 381+2del1 | 381+2del	N/A	10	89473069	89473069
Bannayan-Riley-Ruvalcaba syndrome	PTEN	802-3dupT | 802-3dup1 | 802-3dup	N/A	10	89720648	89720649
Thoracic aortic aneurysms & dissections	ACTA2	124CtoA | H42N	N/A	10	90708564	90708564

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Thoracic aortic aneurysms & dissections	ACTA2	115CtoT | R39C	rs112901682	10	90708573	90708573
Thoracic aortic aneurysms & dissections	ACTA2	112GtoA | G38R	N/A	10	90708576	90708576
Thoracic aortic aneurysms & dissections	ACTA2	76GtoT | D26Y	N/A	10	90708612	90708612
Mohr-Majewski syndrome	TCTN3	566_567delAG | 566_567del2 | 566_567del	N/A	10	97447409	97447410
Hermansky-Pudlak syndrome	HPS1	1996GtoT | E666X	rs121908385	10	100177428	100177428
Hermansky-Pudlak syndrome	HPS1	1691delA | 1691del1 | 1691del	N/A	10	100182178	100182178
Cytochrome c oxidase deficiency	COX15	1030TtoC | S344P	N/A	10	101476176	101476176
Hypogonadotropic hypogonadism, idiopathic	FGF8	118TtoC | F40L	rs137852661	10	103534925	103534925
Kallmann syndrome	FGF8	68AtoT | Q23L	N/A	10	103535498	103535498
Hypogonadotropic hypogonadism, idiopathic	FGF8	40CtoA | H14N	rs137852659	10	103535526	103535526
Hermansky-Pudlak syndrome 6	HPS6	223CtoT | Q75X	N/A	10	103825454	103825454
Medulloblastoma, predisposition to	SUFU	71delC | 71del1 | 71del	N/A	10	104263980	104263980
Medulloblastoma, predisposition to	SUFU	71dupC | 71dup1 | 71dup	N/A	10	104263980	104263981
Medulloblastoma, desmoplastic	SUFU	846dupC | 846dup1 | 846dup	N/A	10	104356986	104356987
Lung cancer, susceptibility to, association with	GPAM	392AtoG | E131G	rs10787428	10	113935379	113935379
Complex I deficiency	GPAM	334GtoA | V112I	N/A	10	113935437	113935437
Gyrate atrophy	OAT	901minus2AtoG	N/A	10	126090410	126090410
Gyrate atrophy	OAT	461GtoT | R154L	rs121965039	10	126097170	126097170
Porphyria, erythropoietic	UROS	217TtoC | C73R	rs121908012	10	127503630	127503630
Altered function	UTF1	217GtoA | G73R	rs11599284	10	135044009	135044009
Creutzfeldt-Jakob disease, variant	SPRN	136dupG | 136dup1 | 136dup	N/A	10	135237049	135237050
Diabetes, type 1	INS	163CtoT | R55C	rs121908261	11	2182039	2182039
Diabetes, neonatal	INS	149AtoG | Y50C	N/A	11	2182053	2182053
Hyperproinsulinaemia	INS	147CtoG | F49L	N/A	11	2182055	2182055
Diabetes, MODY	INS	143TtoC | F48S	rs80356668	11	2182059	2182059
Diabetes, permanent neonatal	INS	143TtoG | F48C	rs80356668	11	2182059	2182059
Diabetes, permanent neonatal	INS	140GtoT | G47V	rs80356667	11	2182062	2182062
Diabetes, MODY	INS	137GtoA | R46Q	rs121908260	11	2182065	2182065
Diabetes, permanent neonatal	INS	71CtoA | A24D	rs80356663	11	2182131	2182131
Diabetes, permanent neonatal	INS	67GtoT | A23S	N/A	11	2182135	2182135
Tyrosine hydroxylase deficiency	TH	1436TtoA | L479Q	N/A	11	2185521	2185521
Dystonia, L-DOPA-responsive	TH	1400AtoG | D467G	N/A	11	2185557	2185557
Parkinsonism, L-DOPA-responsive	TH	1388CtoT | T463M	rs45471299	11	2185569	2185569
Segawa syndrome	TH	1382CtoG | P461R	N/A	11	2185575	2185575
Tyrosine hydroxylase deficiency	TH	1382CtoT | P461L	N/A	11	2185575	2185575
Dystonia, L-DOPA-responsive	TH	1034CtoT | A345V	N/A	11	2187723	2187723
Hypokinetic-hypotonic syndrome, infantile	TH	1032CtoG | F344L	N/A	11	2187725	2187725
Tyrosine hydroxylase deficiency	TH	983GtoT | C328F	rs121917765	11	2187774	2187774
Parkinsonism, L-DOPA-responsive	TH	643CtoT | H215Y	N/A	11	2189097	2189097
Tyrosine hydroxylase deficiency	TH	635AtoC | Q212P	N/A	11	2189105	2189105
Tyrosine hydroxylase deficiency	TH	628GtoA | A210T	N/A	11	2189112	2189112
Dystonia, L-DOPA-responsive	TH	605GtoA | R202H	rs80338892	11	2189135	2189135
Tyrosine hydroxylase deficiency	TH	601CtoT | Q201X	N/A	11	2189139	2189139
Long QT syndrome	KCNQ1	1AtoG | M1V	N/A	11	2466329	2466329
Jervell and Lange-Nielsen syndrome	KCNQ1	2TtoC | M1T	N/A	11	2466330	2466330
Long QT syndrome	KCNQ1	5CtoT | A2V	N/A	11	2466333	2466333
Long QT syndrome & atrial fibrillation	KCNQ1	19CtoT | P7S	N/A	11	2466347	2466347
Atrial fibrillation, susceptibility to	KCNQ1	40CtoT | R14C	N/A	11	2466368	2466368
Long QT syndrome	KCNQ1	107dupT | 107dup1 | 107dup	N/A	11	2466435	2466436
Long QT syndrome	KCNQ1	136GtoA | A46T	N/A	11	2466464	2466464
Long QT syndrome	KCNQ1	151dupT | 151dup1 | 151dup	N/A	11	2466479	2466480
Long QT syndrome	KCNQ1	1343CtoG | P448R	rs12720449	11	2610034	2610034
Long QT syndrome	KCNQ1	1343CtoT | P448L	N/A	11	2610034	2610034
Long QT syndrome	KCNQ1	1343delC | 1343del1 | 1343del	N/A	11	2610034	2610034
Long QT syndrome	KCNQ1	1343dupC | 1343dup1 | 1343dup	N/A	11	2610034	2610035
Long QT syndrome	KCNQ1	1345dupG | 1345dup1 | 1345dup	N/A	11	2610036	2610037
Long QT syndrome	KCNQ1	1643GtoA | G548D	N/A	11	2797242	2797242
Long QT syndrome	KCNQ1	1661TtoC | V554A	N/A	11	2797260	2797260
Long QT syndrome	KCNQ1	1663CtoA | R555S	N/A	11	2797262	2797262
Long QT syndrome	KCNQ1	1663CtoT | R555C	rs120074185	11	2797262	2797262
Long QT syndrome	KCNQ1	1664GtoA | R555H	N/A	11	2797263	2797263
Long QT syndrome	KCNQ1	1669AtoG | K557E	N/A	11	2797268	2797268
Niemann-Pick disease	SMPD1	395TtoC | V132A	N/A	11	6412690	6412690
Niemann-Pick disease	SMPD1	416TtoC | L139P	N/A	11	6412711	6412711
Niemann-Pick disease	SMPD1	502GtoA | G168R	N/A	11	6412797	6412797
Niemann-Pick disease	SMPD1	509GtoA | W170X	N/A	11	6412804	6412804
Niemann-Pick disease	SMPD1	511delG | 511del1 | 511del	N/A	11	6412806	6412806
Niemann-Pick disease	SMPD1	518dupT | 518dup1 | 518dup	N/A	11	6412813	6412814
Niemann-Pick disease	SMPD1	528GtoA | W176X	N/A	11	6412823	6412823
Niemann-Pick disease	SMPD1	533TtoA | I178N	N/A	11	6412828	6412828
Niemann-Pick disease	SMPD1	538_539delTT | 538_539del2 | 538_539del	N/A	11	6412833	6412834
Niemann-Pick disease	SMPD1	557CtoT | P186L	N/A	11	6412852	6412852
Niemann-Pick disease	SMPD1	572dupC | 572dup1 | 572dup	N/A	11	6412867	6412868
Niemann-Pick disease	SMPD1	573delT | 573del1 | 573del	N/A	11	6412868	6412868
Niemann-Pick disease	SMPD1	581delC | 581del1 | 581del	N/A	11	6412876	6412876
Niemann-Pick disease	SMPD1	581dupC | 581dup1 | 581dup	N/A	11	6412876	6412877
Neuronal ceroid lipofuscinosis, late infantile	TPP1	1027GtoA | E343K	rs121908197	11	6637594	6637594
Neuronal ceroid lipofuscinosis, late infantile	TPP1	1016GtoA | R339Q	N/A	11	6637605	6637605
Neuronal ceroid lipofuscinosis, late infantile	TPP1	987_989delGGAinsCTC | 987_989del3ins3 | 987_989delins	N/A	11	6637632	6637634
Neuronal ceroid lipofuscinosis, late infantile	TPP1	406_409dupGCTG | 406_409dup4 | 406_409dup	N/A	11	6638630	6638631
Adenosine monophosphate deaminase deficie	AMPD3	1300CtoT | Q434X	N/A	11	10517150	10517150
Diabetes, permanent neonatal	ABCC8	3763GtoA | G1255S	N/A	11	17419335	17419335
Hyperinsulinism	ABCC8	3521GtoT | C1174F	N/A	11	17426095	17426095
Hyperinsulinism	ABCC8	2064GtoA | W688X	N/A	11	17449466	17449466
Diabetes, permanent neonatal	ABCC8	2062TtoC | W688R	N/A	11	17449468	17449468
Hyperinsulinism	ABCC8	2057TtoC | F686S	N/A	11	17449473	17449473
Hyperinsulinism	ABCC8	1773CtoG | F591L	rs72559726	11	17452405	17452405
Diabetes, NIDDM	ABCC8	1678GtoA | V560M	rs4148619	11	17452500	17452500
Hyperinsulinism	ABCC8	1289GtoA | W430X	N/A	11	17470106	17470106
Hypoglycaemia, persistent hyperinsulinaemic	ABCC8	1252TtoC | C418R	rs67254669	11	17470143	17470143
Deafness, non-syndromic	ush1cpst	1823CtoG | P608R	rs41282932	11	17531093	17531093
Muscular dystrophy, limb girdle 2L	ANO5	1643CtoT | T548I	N/A	11	22283687	22283687
Aniridia	PAX6	1032plus3AtoT	N/A	11	31814983	31814983
Aniridia	PAX6	1032plus2TtoA	N/A	11	31814984	31814984
Aniridia	PAX6	1032plus1GtoA	N/A	11	31814985	31814985

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Aniridia	PAX6	1031AtoG	N/A	11	31814987	31814987
Aniridia	PAX6	1016_1019delATAA | 1016_1019del4 | 1016_1019del	N/A	11	31814999	31815002
Aniridia	PAX6	1017delT | 1017del1 | 1017del	N/A	11	31815001	31815001
Aniridia	PAX6	1000_1001insTGGCATATAACCT | 1000_1001ins13 | 1000_1001ins	N/A	11	31815017	31815018
Aniridia	PAX6	781CtoT | R261X	N/A	11	31815335	31815335
Aniridia	PAX6	775dupT | 775dup1 | 775dup	N/A	11	31815340	31815341
Optic-nerve malformations	PAX6	773TtoC | F258S	rs121907925	11	31815343	31815343
Denys-Drash syndrome	WT1	1334AtoG | H445R	N/A	11	32414217	32414217
Denys-Drash syndrome	WT1	1334AtoT | H445L	N/A	11	32414217	32414217
Denys-Drash syndrome	WT1	1333CtoA | H445N	N/A	11	32414218	32414218
Denys-Drash syndrome	WT1	1333CtoT | H445Y	N/A	11	32414218	32414218
Nephrotic syndrome	WT1	1327_1332delAGGAGA | 1327_1332del6 | 1327_1332del	N/A	11	32414219	32414224
Denys-Drash syndrome	WT1	1323CtoG | H441Q	N/A	11	32414228	32414228
Denys-Drash syndrome	WT1	1321CtoT | H441Y	N/A	11	32414230	32414230
Diffuse mesangial sclerosis	WT1	1311GtoC | Q437H	N/A	11	32414240	32414240
Denys-Drash syndrome	WT1	1311GtoT | Q437H	N/A	11	32414240	32414240
Denys-Drash syndrome	WT1	1310AtoC | Q437P	N/A	11	32414241	32414241
Nephrotic syndrome	WT1	1310AtoG | Q437R	N/A	11	32414241	32414241
Wilms tumour	WT1	1309CtoT | Q437X	N/A	11	32414242	32414242
Denys-Drash syndrome	WT1	1301GtoA | R434H	N/A	11	32414250	32414250
Denys-Drash syndrome	WT1	1301GtoC | R434P	N/A	11	32414250	32414250
Denys-Drash syndrome	WT1	1301GtoT | R434L	N/A	11	32414250	32414250
Denys-Drash syndrome	WT1	1300CtoT | R434C	N/A	11	32414251	32414251
Nephrotic syndrome	WT1	1294TtoC | F432L	N/A	11	32414257	32414257
Wilms tumour	WT1	962GtoC | G321A	N/A	11	32438075	32438075
Multiple exostoses	EXT2	1234CtoT | Q412X	N/A	11	44193221	44193221
Multiple osteochondromas	EXT2	1250GtoC | R417P	N/A	11	44193237	44193237
Multiple exostoses	EXT2	1251delG | 1251del1 | 1251del	N/A	11	44193238	44193238
Multiple exostoses	EXT2	1257TtoA | Y419X	N/A	11	44193244	44193244
Multiple exostoses	EXT2	1263TtoA | Y421X	N/A	11	44193250	44193250
Multiple exostoses	EXT2	1262_1263dupAT | 1262_1263dup2 | 1262_1263dup	N/A	11	44193250	44193251
Peroxisome biogenesis disorder	PEX16	992AtoG | Y331C	N/A	11	45931824	45931824
Peroxisome biogenesis disorder	PEX16	984delG | 984del1 | 984del	N/A	11	45931832	45931832
Prothrombin deficiency	F2	175GtoC | E59Q	N/A	11	46741347	46741347
Richter syndrome, increased risk, association v	LRP4	4660GtoA | G1554S	rs2306029	11	46893108	46893108
Hip bone mineral density, association with	LRP4	3256AtoG | I1086V	rs6485702	11	46898771	46898771
Cardiomyopathy, hypertrophic	MYBPC3	3797GtoA | C1266Y	N/A	11	47353640	47353640
Cardiomyopathy, hypertrophic	MYBPC3	3796TtoC | C1266R	N/A	11	47353641	47353641
Cardiomyopathy, hypertrophic	MYBPC3	3794AtoT | E1265V	N/A	11	47353643	47353643
Cardiomyopathy, dilated	MYBPC3	3791GtoT | C1264F	N/A	11	47353646	47353646
Cardiomyopathy, hypertrophic	MYBPC3	3259AtoG | K1087E	N/A	11	47354816	47354816
Cardiomyopathy, hypertrophic	MYBPC3	3258dupG | 3258dup1 | 3258dup	N/A	11	47354816	47354817
Cardiomyopathy, hypertrophic	MYBPC3	3257_3258dupGG | 3257_3258dup2 | 3257_3258dup	N/A	11	47354816	47354817
Cardiomyopathy, hypertrophic	MYBPC3	3251TtoC | L1084P	N/A	11	47354824	47354824
Cardiomyopathy, hypertrophic	MYBPC3	2690_2699delGAGCAGGAGG | 2690_2699del10 | 2690_2699del	N/A	11	47357466	47357475
Cardiomyopathy, hypertrophic	MYBPC3	2686GtoA | V896M	rs35078470	11	47357479	47357479
Cardiomyopathy, hypertrophic	MYBPC3	2670dupG | 2670dup1 | 2670dup	N/A	11	47357494	47357495
Cardiomyopathy, hypertrophic	MYBPC3	2670GtoA | W890X	N/A	11	47357495	47357495
Cardiomyopathy, hypertrophic	MYBPC3	2654CtoT | T885M	N/A	11	47357511	47357511
Cardiomyopathy, hypertrophic	MYBPC3	2573GtoA | S858N	N/A	11	47358971	47358971
Cardiomyopathy, hypertrophic	MYBPC3	2559CtoT | G853G	N/A	11	47358985	47358985
Cardiomyopathy, hypertrophic	MYBPC3	655GtoC | V219L	N/A	11	47370092	47370092
Cardiomyopathy, hypertrophic	MYBPC3	655GtoT | V219F	N/A	11	47370092	47370092
Cardiomyopathy, hypertrophic	MYBPC3	655minus1GtoA	N/A	11	47370093	47370093
Cardiomyopathy, hypertrophic	MYBPC3	655minus2AtoC	N/A	11	47370094	47370094
Cardiomyopathy, hypertrophic	MYBPC3	418GtoC | A140P	N/A	11	47371652	47371652
Cardiomyopathy, hypertrophic	MYBPC3	343GtoT | G115X	N/A	11	47372116	47372116
Angioneurotic oedema	SERPING1	minus22minus2AtoG	N/A	11	57365720	57365720
Angioneurotic oedema	SERPING1	minus22minus1GtoA	N/A	11	57365721	57365721
Adult vitelliform macular dystrophy	BEST1	250TtoG | F84V	N/A	11	61723192	61723192
Best macular dystrophy	BEST1	253TtoC | Y85H	rs28940274	11	61723195	61723195
Nanophthalmos and retinal dystrophy	BEST1	256GtoA | V86M	rs121918289	11	61723198	61723198
Best macular dystrophy	BEST1	266TtoC | V89A	N/A	11	61723208	61723208
Bestrophinopathy	BEST1	266_282delTCGTGACCCGCTGGTGG | 266_282del17 | 266_282del	N/A	11	61723208	61723224
Best macular dystrophy	BEST1	272CtoT | T91I	N/A	11	61723214	61723214
Best macular dystrophy	BEST1	274CtoA | R92S	N/A	11	61723216	61723216
Adult vitelliform macular dystrophy	BEST1	274CtoG | R92G	N/A	11	61723216	61723216
Best macular dystrophy	BEST1	274CtoT | R92C	N/A	11	61723216	61723216
Best macular dystrophy	BEST1	275GtoA | R92H	N/A	11	61723217	61723217
Adult vitelliform macular dystrophy	BEST1	277TtoC | W93R	N/A	11	61723219	61723219
Best macular dystrophy	BEST1	279GtoC | W93C	rs28940273	11	61723221	61723221
Adult vitelliform macular dystrophy	BEST1	286CtoG | Q96E	N/A	11	61723228	61723228
Best macular dystrophy	BEST1	287AtoG | Q96R	N/A	11	61723229	61723229
Best macular dystrophy	BEST1	288GtoC | Q96H	N/A	11	61723230	61723230
Best macular dystrophy	BEST1	295AtoT | N99Y	N/A	11	61723237	61723237
Best macular dystrophy	BEST1	297CtoA | N99K	N/A	11	61723239	61723239
Best macular dystrophy	BEST1	299TtoG | L100R	N/A	11	61723241	61723241
Best macular dystrophy	BEST1	301CtoA | P101T	N/A	11	61723243	61723243
Adult vitelliform macular dystrophy	BEST1	302CtoT | P101L	N/A	11	61723244	61723244
Best macular dystrophy	BEST1	304TtoC | W102R	N/A	11	61723246	61723246
Gout, primary	SLC22A12	889CtoG | Q297E	N/A	11	64366046	64366046
Hypouricaemia, renal	SLC22A12	889CtoT | Q297X	N/A	11	64366046	64366046
Hypouricaemia, renal	SLC22A12	894GtoT | E298D	rs121907894	11	64366051	64366051
Gout, primary	SLC22A12	914TtoG | I305S	N/A	11	64366071	64366071
McArdle disease	PYGM	1768plus1GtoA	N/A	11	64519395	64519395
McArdle disease	PYGM	1760TtoC | L587P	N/A	11	64519404	64519404
McArdle disease	PYGM	1628AtoC | K543T	rs119103252	11	64519536	64519536
McArdle disease	PYGM	1627AtoT | K543X	N/A	11	64519537	64519537
McArdle disease	PYGM	1621GtoT | E541X	rs119103257	11	64519543	64519543
Hyperparathyroidism	MEN1	830CtoA | P277H	N/A	11	64574565	64574565
Multiple endocrine neoplasia 1	MEN1	828CtoA | Y276X	N/A	11	64574567	64574567
Multiple endocrine neoplasia 1	MEN1	825minus1GtoC	N/A	11	64574571	64574571
Multiple endocrine neoplasia 1	MEN1	825minus2AtoG	N/A	11	64574572	64574572
Multiple endocrine neoplasia 1	MEN1	738_741delCATTinsAGCGAGTCG | 738_741del4ins9 | 738_741delins	N/A	11	64575066	64575069
Hyperparathyroidism	MEN1	734delC | 734del1 | 734del	N/A	11	64575073	64575073

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Multiple endocrine neoplasia 1	MEN1	594GtoA | W198X	rs104894257	11	64575423	64575423
Multiple endocrine neoplasia 1	MEN1	593GtoA | W198X	rs104894258	11	64575424	64575424
Multiple endocrine neoplasia 1	MEN1	589_591delACC | 589_591del3 | 589_591del	N/A	11	64575426	64575428
Hyperparathyroidism	MEN1	590CtoT | T197I	N/A	11	64575427	64575427
Multiple endocrine neoplasia 1	MEN1	584AtoG | E195G	N/A	11	64575433	64575433
Hyperparathyroidism and pancreatic neoplasia	MEN1	564_581delCAATGGGGAGCAGACAGC | 564_581del18 | 564_581del	N/A	11	64575436	64575453
Multiple endocrine neoplasia 1	MEN1	578CtoT | T193I	N/A	11	64575439	64575439
Hyperparathyroidism	MEN1	574CtoT | Q192X	N/A	11	64575443	64575443
Multiple endocrine neoplasia 1	MEN1	570_571dupGG | 570_571dup2 | 570_571dup	N/A	11	64575445	64575446
Multiple endocrine neoplasia 1	MEN1	571GtoT | E191X	N/A	11	64575446	64575446
Multiple endocrine neoplasia 1	MEN1	571delG | 571del1 | 571del	N/A	11	64575446	64575446
Hyperparathyroidism	MEN1	570_571delGGinsC | 570_571del2ins1 | 570_571delins	N/A	11	64575446	64575447
Multiple endocrine neoplasia 1	MEN1	563_564delCC | 563_564del2 | 563_564del	N/A	11	64575453	64575454
Hyperparathyroidism	MEN1	563CtoT | P188L	N/A	11	64575454	64575454
Multiple endocrine neoplasia 1	MEN1	558delT | 558del1 | 558del	N/A	11	64575459	64575459
Hyperparathyroidism	MEN1	551TtoA | V184E	rs104894262	11	64575466	64575466
Multiple endocrine neoplasia 1	MEN1	549GtoA | W183X	N/A	11	64575468	64575468
Multiple endocrine neoplasia 1	MEN1	549GtoC | W183C	N/A	11	64575468	64575468
Multiple endocrine neoplasia 1	MEN1	548GtoA | W183X	N/A	11	64575469	64575469
Multiple endocrine neoplasia 1	MEN1	548GtoC | W183S	N/A	11	64575469	64575469
Multiple endocrine neoplasia 1	MEN1	547TtoC | W183R	N/A	11	64575470	64575470
Multiple endocrine neoplasia 1	MEN1	545_546insAAACACTACTA | 545_546ins11 | 545_546ins	N/A	11	64575471	64575472
Multiple endocrine neoplasia 1	MEN1	542AtoG | H181R	N/A	11	64575475	64575475
Multiple endocrine neoplasia 1	MEN1	537GtoT | E179D	N/A	11	64575480	64575480
Multiple endocrine neoplasia 1	MEN1	535GtoA | E179K	N/A	11	64575482	64575482
Multiple endocrine neoplasia 1	MEN1	535GtoC | E179Q	N/A	11	64575482	64575482
Hyperparathyroidism	MEN1	532_535delTCTG | 532_535del4 | 532_535del	N/A	11	64575482	64575485
Multiple endocrine neoplasia 1	MEN1	530TtoC | L177P	N/A	11	64575487	64575487
Multiple endocrine neoplasia 1	MEN1	526GtoC | A176P	N/A	11	64575491	64575491
Multiple endocrine neoplasia 1	MEN1	526delG | 526del1 | 526del	N/A	11	64575491	64575491
Multiple endocrine neoplasia 1	MEN1	524TtoG | L175R	N/A	11	64575493	64575493
Multiple endocrine neoplasia 1	MEN1	523delC | 523del1 | 523del	N/A	11	64575494	64575494
Multiple endocrine neoplasia 1	MEN1	514GtoT | D172Y	N/A	11	64575503	64575503
Pyruvate carboxylase deficiency	PC	1023minus1GtoT	N/A	11	66633821	66633821
Cold-induced sweating syndrome	CLCF1	321CtoA | Y107X	rs104894198	11	67132964	67132964
Pituitary adenoma	AIP	280minus1GtoC	N/A	11	67256737	67256737
Pituitary adenoma	AIP	286_287delGT | 286_287del2 | 286_287del	N/A	11	67256744	67256745
Pituitary adenoma	AIP	308AtoG | K103R	N/A	11	67256766	67256766
Pituitary adenoma	AIP	338_341dupACCC | 338_341dup4 | 338_341dup	N/A	11	67256799	67256800
Acromegaly	AIP	350delG | 350del1 | 350del	N/A	11	67256808	67256808
Pituitary adenoma	AIP	752delT | 752del1 | 752del	N/A	11	67257893	67257893
Pituitary adenoma	AIP	769AtoG | I257V	N/A	11	67257910	67257910
Osteopetrosis, infantile malignant	TCIRG1	242delC | 242del1 | 242del	N/A	11	67810155	67810155
Osteopetrosis, infantile malignant	TCIRG1	831_836delCCTGAGinsA | 831_836del6ins1 | 831_836delins	N/A	11	67811622	67811627
Osteoporosis-pseudoglioma syndrome	LRP5	29GtoA | W10X	rs121908660	11	68080211	68080211
Osteoporosis-pseudoglioma syndrome	LRP5	43_60delCTGCTGCTGCTGCTGCTG | 43_60del18 | 43_60del	N/A	11	68080225	68080242
Osteoporosis-pseudoglioma syndrome	LRP5	920CtoT | S307F	N/A	11	68133075	68133075
Osteoporosis-pseudoglioma syndrome	LRP5	1000_1004dupAGGAC | 1000_1004dup5 | 1000_1004dup	N/A	11	68133159	68133160
Osteoporosis-pseudoglioma syndrome	LRP5	1015plus1GtoT	N/A	11	68133171	68133171
Spinal muscular atrophy with respiratory distr	IGHMBP2	50TtoC | L17P	N/A	11	68671470	68671470
Spinal muscular atrophy with respiratory distr	IGHMBP2	1648dupC | 1648dup1 | 1648dup	N/A	11	68702782	68702783
Deafness, non-syndromic	LRTOMT	242GtoA | R81Q	rs137853185	11	71817140	71817140
Obesity, severe, early-onset	UCP3	754CtoT | Q252X	N/A	11	73714942	73714942
Usher syndrome 1b	MYO7A	47TtoA | L16X	N/A	11	76853783	76853783
Usher syndrome 1	MYO7A	52CtoT | Q18X	N/A	11	76853788	76853788
Usher syndrome 1b	MYO7A	1884CtoA | C628X	rs121965083	11	76883880	76883880
Usher syndrome 1b	MYO7A	1900CtoT | R634X	rs111033180	11	76883896	76883896
Retinitis pigmentosa, mild & deafness	MYO7A	1935GtoA	N/A	11	76883931	76883931
Usher syndrome 1b	MYO7A	1935plus1GtoA	N/A	11	76883932	76883932
Deafness, non-syndromic, autosomal dominar	MYO7A	2662_2670delAAGGCCAAG | 2662_2670del9 | 2662_2670del	N/A	11	76891495	76891503
Usher syndrome 1	MYO7A	2663dupA | 2663dup1 | 2663dup	N/A	11	76891496	76891497
Usher syndrome 1	MYO7A	5215CtoT | R1739X	N/A	11	76914151	76914151
Usher syndrome 1b	MYO7A	5227CtoT | R1743W	rs111033287	11	76914163	76914163
Usher syndrome 1b	MYO7A	5309CtoA | A1770D	N/A	11	76914245	76914245
Usher syndrome 1b	MYO7A	6560GtoA | G2187D	N/A	11	76925653	76925653
Metaphyseal anadysplasia	MMP13	694CtoA | H232N	rs121909500	11	102822846	102822846
Acetoacetyl-CoA-thiolase deficiency	ACAT1	52dupC | 52dup1 | 52dup	N/A	11	107992385	107992386
Myopathy, myofibrillar, infantile-onset	CRYAB	343delT | 343del1 | 343del	N/A	11	111779673	111779673
Myofibrillar myopathy, cardiomyopathy & pos	CRYAB	325GtoC | D109H	N/A	11	111779691	111779691
Porphyria, acute intermittent	HMBS	34minus21AtoG	N/A	11	118958944	118958944
Porphyria, acute intermittent	HMBS	34minus2AtoT	N/A	11	118958963	118958963
Porphyria, acute intermittent	HMBS	34minus1GtoT	N/A	11	118958964	118958964
Porphyria, acute intermittent	HMBS	34GtoT | E12X	N/A	11	118958965	118958965
Porphyria, acute intermittent	HMBS	613minus3CtoA	N/A	11	118962832	118962832
Porphyria, acute intermittent	HMBS	613minus2AtoG	N/A	11	118962833	118962833
Porphyria, acute intermittent	HMBS	613minus1GtoA	N/A	11	118962834	118962834
Porphyria, acute intermittent	HMBS	613minus1GtoC	N/A	11	118962834	118962834
Porphyria, acute intermittent	HMBS	613minus1GtoT	N/A	11	118962834	118962834
Porphyria, acute intermittent	HMBS	625GtoA | E209K	N/A	11	118962847	118962847
Porphyria, acute intermittent	HMBS	630delA | 630del1 | 630del	N/A	11	118962852	118962852
Porphyria, acute intermittent	HMBS	634AtoG | M212V	N/A	11	118962856	118962856
Porphyria, acute intermittent	HMBS	635TtoG | M212R	N/A	11	118962857	118962857
Porphyria, acute intermittent	HMBS	639TtoG | Y213X	N/A	11	118962861	118962861
Porphyria, acute intermittent	HMBS	643GtoA | V215M	N/A	11	118962865	118962865
Porphyria, acute intermittent	HMBS	646_647insA | 646_647ins1 | 646_647ins	N/A	11	118962868	118962869
Porphyria, acute intermittent	HMBS	647GtoA | G216D	rs118204116	11	118962869	118962869
Porphyria, acute intermittent	HMBS	650AtoG | Q217R	N/A	11	118962872	118962872
Porphyria, acute intermittent	HMBS	650AtoT | Q217L	N/A	11	118962872	118962872
Porphyria, acute intermittent	HMBS	651GtoC | Q217H	N/A	11	118962873	118962873
Porphyria, acute intermittent	HMBS	651GtoT | Q217H	N/A	11	118962873	118962873
Porphyria, acute intermittent	HMBS	651plus1GtoC	N/A	11	118962874	118962874
Porphyria, acute intermittent	HMBS	651plus2TtoC	N/A	11	118962875	118962875
Limb-girdle congenital myasthenic syndrome v	DPAGT1	698dupT | 698dup1 | 698dup	N/A	11	118969142	118969143
Limb-girdle congenital myasthenic syndrome v	DPAGT1	478GtoA | G160S	N/A	11	118971358	118971358
Nanophthalmos hyperopia	MFRP	1150dupC | 1150dup1 | 1150dup	N/A	11	119213687	119213688

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Nanophthalmos-retinitis pigmentosa complex	MFRP	951CtoA | Y317X	N/A	11	119215049	119215049
Gaze palsy, horizontal, with progressive scolio	ROBO3	2113TtoC | S705P	rs121918272	11	124745046	124745046
Megalencephalic leukoencephalopathy with s	HEPACAM	461_462delCA | 461_462del2 | 461_462del	N/A	11	124793872	124793873
Cardiac arrhythmia	KCNA5	98AtoT | E33V	rs71584818	12	5153411	5153411
Von Willebrand disease 1	VWF	6599minus20AtoT	rs61750621	12	6101204	6101204
Von Willebrand disease 1	VWF	6536CtoT | S2179F	rs61750620	12	6103090	6103090
Von Willebrand disease 3	VWF	6520TtoG | C2174G	rs61750619	12	6103106	6103106
Von Willebrand disease 2a	VWF	6254GtoA | C2085Y	N/A	12	6103583	6103583
Von Willebrand disease 3	VWF	6197AtoG | N2066S	N/A	12	6103640	6103640
Von Willebrand disease	VWF	4946TtoA | I1649N	N/A	12	6127638	6127638
Von Willebrand disease 1	VWF	4944delT | 4944del1 | 4944del	rs61750591	12	6127640	6127640
Von Willebrand disease 2a	VWF	4942CtoT | P1648S	rs61750590	12	6127642	6127642
Increased protein cleavage by ADAMTS13	VWF	4927GtoA | G1643S	N/A	12	6127657	6127657
Von Willebrand disease 2n	VWF	2649_2650insTTTG | 2649_2650ins4 | 2649_2650ins	N/A	12	6143889	6143890
Von Willebrand disease 1	VWF	2642TtoG | L881R	N/A	12	6143897	6143897
Von Willebrand, Normandy variant	VWF	2635GtoA | D879N	rs61748485	12	6143904	6143904
Von Willebrand disease 3	VWF	2574_2584delCACAGACCATG | 2574_2584del11 | 2574_2584del	N/A	12	6143955	6143965
Von Willebrand disease 1	VWF	2574CtoG | C858W	rs184227165	12	6143965	6143965
Von Willebrand, Normandy variant	VWF	2573GtoT | C858F	rs61748484	12	6143966	6143966
Von Willebrand, Normandy variant	VWF	2561GtoA | R854Q	rs41276738	12	6143978	6143978
Von Willebrand disease 2	VWF	2560CtoT | R854W	N/A	12	6143979	6143979
Reduced vWF plasma protein levels	VWF	2555GtoA | R852Q	rs216321	12	6143984	6143984
Von Willebrand disease 3	VWF	1071CtoA | Y357X	rs61754002	12	6181535	6181535
Periodic fever, autosomal dominant	TNFRSF1A	304TtoC | C102R	rs104895236	12	6442921	6442921
Pseudohypoaldosteronism 1	SCNN1A	1356delC | 1356del1 | 1356del	N/A	12	6463608	6463608
Autism	NCAPD2	1706TtoC | I569T	N/A	12	6630268	6630268
Triosephosphate isomerase deficiency	TPI1	32dupG | 32dup1 | 32dup	N/A	12	6976762	6976763
Triosephosphate isomerase deficiency	TPI1	86_87delTG | 86_87del2 | 86_87del	N/A	12	6976816	6976817
Triosephosphate isomerase deficiency	TPI1	436GtoT | E146X	rs121964850	12	6978533	6978533
Alzheimer disease, association with	A2M	2597minus4TtoA	rs11609582	12	9242623	9242623
Keutel syndrome	MGP	61plus1GtoA	rs111798215	12	15038664	15038664
Keutel syndrome	MGP	43delG | 43del1 | 43del	N/A	12	15038683	15038683
Sudden unexpeted death with negative autop	PKP2	1618GtoA | E540K	N/A	12	32994032	32994032
Arrhythmogenic right ventricular dysplasia	PKP2	1613GtoA | W538X	N/A	12	32994037	32994037
Arrhythmogenic right ventricular dysplasia	PKP2	1597_1600delATCC | 1597_1600del4 | 1597_1600del	N/A	12	32994050	32994053
Arrhythmogenic right ventricular dysplasia	PKP2	1271TtoC | F424S	N/A	12	33003807	33003807
Arrhythmogenic right ventricular dysplasia/ca	PKP2	1256TtoC | L419S	N/A	12	33003822	33003822
Arrhythmogenic right ventricular dysplasia	PKP2	1237CtoT | R413X	N/A	12	33003841	33003841
Arrhythmogenic right ventricular dysplasia/ca	PKP2	1231dupG | 1231dup1 | 1231dup	N/A	12	33003846	33003847
Arrhythmogenic right ventricular dysplasia	PKP2	1211dupT | 1211dup1 | 1211dup	N/A	12	33003866	33003867
Arrhythmogenic right ventricular cardiomyopa	PKP2	1202_1209delTGCAGCTC | 1202_1209del8 | 1202_1209del	N/A	12	33003869	33003876
Arrhythmogenic right ventricular dysplasia/ca	PKP2	1205delA | 1205del1 | 1205del	N/A	12	33003873	33003873
Arrhythmogenic right ventricular dysplasia	PKP2	1171minus2AtoG	N/A	12	33003909	33003909
Parkinson disease, reduced risk, association w	LRRK2	4193GtoA | R1398H	rs7133914	12	40702911	40702911
Scott syndrome	ANO6	1219dupT | 1219dup1 | 1219dup	N/A	12	45781997	45781998
Rickets, vitamin D resistant, without alopecia	VDR	856TtoC | W286R	N/A	12	48240491	48240491
Rickets, vitamin D resistant	VDR	570TtoG | C190W	N/A	12	48250925	48250925
Stickler syndrome	COL2A1	3392GtoC | G1131A	N/A	12	48370638	48370638
Kniest dysplasia	COL2A1	3383GtoT | G1128V	N/A	12	48370647	48370647
Spondyloepiphyseal dysplasia	COL2A1	3364GtoA | G1122R	N/A	12	48370666	48370666
Hypochondrogenesis	COL2A1	3337GtoT | G1113C	N/A	12	48370693	48370693
Hypochondrogenesis	COL2A1	3062_3079delCCGTGGGTCCTCCTGGCC | 3062_3079del18 | 3062_3079del	N/A	12	48371825	48371842
Spondyloepiphyseal dysplasia	COL2A1	3056_3058delCTG | 3056_3058del3 | 3056_3058del	N/A	12	48371846	48371848
Achondrogenesis 2	COL2A1	3050GtoT | G1017V	N/A	12	48371854	48371854
Stickler syndrome	COL2A1	2839CtoT | Q947X	N/A	12	48372436	48372436
Stickler syndrome	COL2A1	2814delT | 2814del1 | 2814del	N/A	12	48372461	48372461
Stickler syndrome	COL2A1	2813dupC | 2813dup1 | 2813dup	N/A	12	48372461	48372462
Stickler syndrome	COL2A1	2813delC | 2813del1 | 2813del	N/A	12	48372462	48372462
Hypochondrogenesis	COL2A1	2807GtoA | G936D	N/A	12	48372468	48372468
Spondyloepimetaphyseal dysplasia, Strudwick	COL2A1	2725GtoT | G909C	rs121912875	12	48373302	48373302
Stickler syndrome	COL2A1	2723dupC | 2723dup1 | 2723dup	N/A	12	48373303	48373304
Stickler syndrome	COL2A1	2715dupT | 2715dup1 | 2715dup	N/A	12	48373311	48373312
Stickler syndrome	COL2A1	2710CtoT | R904C	rs121912882	12	48373317	48373317
Achondrogenesis 2	COL2A1	2681GtoA | G894E	N/A	12	48373346	48373346
Stickler syndrome	COL2A1	2588_2604delCTGGTCCTCAGGGCCCC | 2588_2604del17 | 2588_2604del	N/A	12	48374358	48374374
Spondyloepiphyseal dysplasia	COL2A1	2463+4_2463+7delAGTG | 2463+4_2463+7del4 | 2463+4_2463+7del	N/A	12	48375119	48375122
Stickler syndrome	COL2A1	2436dupA | 2436dup1 | 2436dup	N/A	12	48375152	48375153
Stickler syndrome	COL2A1	2116GtoT | E706X	N/A	12	48376708	48376708
Stickler syndrome	COL2A1	2101CtoT | R701X	N/A	12	48376723	48376723
Stickler syndrome	COL2A1	1970delG | 1970del1 | 1970del	N/A	12	48377491	48377491
Stickler syndrome	COL2A1	1962CtoT	N/A	12	48377499	48377499
Stickler syndrome	COL2A1	1957CtoT | R653X	rs121912893	12	48377504	48377504
Stickler syndrome	COL2A1	1943dupG | 1943dup1 | 1943dup	N/A	12	48377517	48377518
Stickler syndrome	COL2A1	1942minus2AtoG	N/A	12	48377521	48377521
Kniest dysplasia	COL2A1	1734plus5GtoA	N/A	12	48379312	48379312
Stickler syndrome	COL2A1	1732delT | 1732del1 | 1732del	N/A	12	48379319	48379319
Stickler syndrome	COL2A1	1714CtoT | Q572X	N/A	12	48379337	48379337
Stickler syndrome	COL2A1	1693CtoT | R565C	rs121912884	12	48379358	48379358
Obesity	WNT10B	767GtoA | C256Y	N/A	12	49360281	49360281
Kabuki syndrome	MLL2	14760_14761insA | 14760_14761ins1 | 14760_14761ins	N/A	12	49420988	49420989
Kabuki syndrome	MLL2	14710CtoT | R4904X	N/A	12	49421039	49421039
Kabuki syndrome	MLL2	8366plus5GtoC	N/A	12	49433000	49433000
Kabuki syndrome	MLL2	5124_5125delAC | 5124_5125del2 | 5124_5125del	N/A	12	49438046	49438047
Polymicrogyria	TUBA1A	13AtoC | I5L	N/A	12	49580607	49580607
Diabetes insipidus, nephrogenic	AQP2	439GtoA | A147T	rs104894334	12	50348016	50348016
Diabetes insipidus, nephrogenic	AQP2	450TtoA | D150E	N/A	12	50348027	50348027
Diabetes insipidus, nephrogenic	AQP2	607minus1GtoA	N/A	12	50349181	50349181
Diabetes insipidus, nephrogenic	AQP2	631GtoC | G211R	N/A	12	50349206	50349206
Diabetes insipidus, nephrogenic	AQP2	643GtoT | G215C	N/A	12	50349218	50349218
Diabetes insipidus, nephrogenic	AQP2	646TtoC | S216P	rs104894329	12	50349221	50349221
Diabetes insipidus, nephrogenic	AQP2	647CtoT | S216F	N/A	12	50349222	50349222
Diabetes insipidus, nephrogenic	AQP2	652delC | 652del1 | 652del	N/A	12	50349227	50349227
Diabetes insipidus, nephrogenic	AQP2	682AtoG | K228E	N/A	12	50349257	50349257
Diabetes insipidus, nephrogenic	AQP2	721delG | 721del1 | 721del	N/A	12	50349296	50349296
Diabetes insipidus, nephrogenic	AQP2	727delG | 727del1 | 727del	N/A	12	50349302	50349302

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Haemorrhagic telangiectasia 2	ACVRL1	536AtoC | D179A	N/A	12	52307768	52307768
Haemorrhagic telangiectasia 2	ACVRL1	540_541insA | 540_541ins1 | 540_541ins	N/A	12	52307772	52307773
Haemorrhagic telangiectasia	ACVRL1	563delC | 563del1 | 563del	N/A	12	52307795	52307795
Haemorrhagic telangiectasia 2	ACVRL1	567delG | 567del1 | 567del	N/A	12	52307799	52307799
Haemorrhagic telangiectasia 2	ACVRL1	573delC | 573del1 | 573del	N/A	12	52307805	52307805
Haemorrhagic telangiectasia 2	ACVRL1	590CtoT | T197I	N/A	12	52307822	52307822
Haemorrhagic telangiectasia	ACVRL1	600dupG | 600dup1 | 600dup	N/A	12	52307832	52307833
Haemorrhagic telangiectasia	ACVRL1	601CtoT | Q201X	N/A	12	52307833	52307833
Haemorrhagic telangiectasia	ACVRL1	602AtoC | Q201P	N/A	12	52307834	52307834
Pulmonary arterial hypertension	ACVRL1	602AtoG | Q201R	N/A	12	52307834	52307834
Haemorrhagic telangiectasia	ACVRL1	611TtoG | L204W	N/A	12	52307843	52307843
Haemorrhagic telangiectasia 2	ACVRL1	617AtoG | E206G	N/A	12	52307849	52307849
Haemorrhagic telangiectasia 2	ACVRL1	623_624dupTG | 623_624dup2 | 623_624dup	N/A	12	52307856	52307857
Haemorrhagic telangiectasia 2	ACVRL1	1048plus1GtoA	N/A	12	52309285	52309285
Monilethrix	KRT86	1237GtoA | E413K	rs121909129	12	52700054	52700054
Monilethrix	KRT86	1239GtoT | E413D	rs121909130	12	52700056	52700056
Pachyonychia congenita	KRT6A	1460minus2AtoC	N/A	12	52881741	52881741
Liver disease, susceptibility to	KRT18	782GtoA | R261Q	rs57354642	12	53345389	53345389
Triple-A syndrome	AAAS	788CtoT | S263L	N/A	12	53703407	53703407
Triple-A syndrome	AAAS	787TtoC | S263P	rs121918550	12	53703408	53703408
Triple-A syndrome	AAAS	771delG | 771del1 | 771del	N/A	12	53703424	53703424
Potential protein deficiency	SMUG1	7CtoT | Q3X	rs2233919	12	54577718	54577718
GnRH deficiency	TAC3	238CtoA | R80S	N/A	12	57407143	57407143
Hypogonadotropic hypogonadism	TAC3	209minus1GtoC	rs146391497	12	57407173	57407173
Alzheimer's disease, association with	LRP1	300CtoT | D100D	rs1799986	12	57535266	57535266
Mucopolysaccharidosis IIId	GNS	911_919delAGAAACTGG | 911_919del9 | 911_919del	N/A	12	65133236	65133244
Mucopolysaccharidosis IIId	GNS	109dupG | 109dup1 | 109dup	N/A	12	65152947	65152948
Mucopolysaccharidosis IIId	GNS	59_66delCCTCCTGC | 59_66del8 | 59_66del	N/A	12	65152991	65152998
Bardet-Biedl syndrome	BBS10	197plus1GtoT	N/A	12	76741941	76741941
Reduced function	APAF1	431AtoC | K144T	N/A	12	99043367	99043367
Mucolipidosis III	GNPTAB	1001GtoA | R334Q	N/A	12	102164296	102164296
Mucolipidosis II	GNPTAB	1001GtoT | R334L	N/A	12	102164296	102164296
Mucolipidosis III	GNPTAB	1000CtoT | R334X	N/A	12	102164297	102164297
Phenylketonuria	PAH	1049CtoA | S350Y	rs62508628	12	103238130	103238130
Phenylketonuria	PAH	1048TtoA | S350T	rs62517183	12	103238131	103238131
Phenylketonuria	PAH	1044_1047dupGTCA | 1044_1047dup4 | 1044_1047dup	N/A	12	103238131	103238132
Phenylketonuria	PAH	1046CtoA | S349X	N/A	12	103238133	103238133
Phenylketonuria	PAH	1046CtoT | S349L	rs62507279	12	103238133	103238133
Phenylketonuria	PAH	1045TtoC | S349P	rs62508646	12	103238134	103238134
Phenylketonuria	PAH	1045TtoG | S349A	rs62508646	12	103238134	103238134
Phenylketonuria	PAH	1038delG | 1038del1 | 1038del	rs62516063	12	103238141	103238141
Phenylketonuria	PAH	1036GtoA | G346R	N/A	12	103238143	103238143
Phenylketonuria	PAH	1036GtoC | G346R	rs62508688	12	103238143	103238143
Phenylketonuria	PAH	1022AtoC | K341T	N/A	12	103238157	103238157
Phenylketonuria	PAH	1022AtoG | K341R	rs62516153	12	103238157	103238157
Phenylketonuria	PAH	1021AtoT | K341X	rs62517200	12	103238158	103238158
Phenylketonuria	PAH	1019TtoC | I340T	rs62508720	12	103238160	103238160
Phenylketonuria	PAH	1012GtoT | D338Y	rs62516150	12	103238167	103238167
Phenylketonuria	PAH	1010GtoT | G337V	rs62517206	12	103238169	103238169
Phenylketonuria	PAH	1007AtoG | Q336R	rs62508735	12	103238172	103238172
Phenylketonuria	PAH	1006CtoT | Q336X	rs62516061	12	103238173	103238173
Phenylketonuria	PAH	1001GtoC | C334S	rs62517174	12	103238178	103238178
Phenylketonuria	PAH	969plus6TtoA	rs62517196	12	103240667	103240667
Hyperphenylalaninaemia	PAH	969plus5GtoA	rs62508637	12	103240668	103240668
Phenylketonuria	PAH	969plus1GtoA	rs62508584	12	103240672	103240672
Increased activity	PAH	969AtoG | T323T	N/A	12	103240673	103240673
Phenylketonuria	PAH	967_969delACA | 967_969del3 | 967_969del	N/A	12	103240673	103240675
Phenylketonuria	PAH	965CtoG | A322G	rs62514958	12	103240677	103240677
Phenylketonuria	PAH	965CtoT | A322V	N/A	12	103240677	103240677
Phenylketonuria	PAH	964GtoA | A322T	rs62514957	12	103240678	103240678
Phenylketonuria	PAH	960GtoC | K320N	N/A	12	103240682	103240682
Phenylketonuria	PAH	953TtoC | I318T	rs62642918	12	103240689	103240689
Phenylketonuria	PAH	949TtoC | Y317H	N/A	12	103240693	103240693
Phenylketonuria	PAH	943GtoT | D315Y	rs62642917	12	103240699	103240699
Phenylketonuria	PAH	941CtoA | P314H	rs62642940	12	103240701	103240701
Phenylketonuria	PAH	941delC | 941del1 | 941del	N/A	12	103240701	103240701
Phenylketonuria	PAH	940CtoA | P314T	N/A	12	103240702	103240702
Phenylketonuria	PAH	940CtoT | P314S	N/A	12	103240702	103240702
Phenylketonuria	PAH	930_940delTCTGGGTGCAC | 930_940del11 | 930_940del	N/A	12	103240702	103240712
Phenylketonuria	PAH	938CtoT | A313V	rs62642914	12	103240704	103240704
Phenylketonuria	PAH	937GtoA | A313T	rs62642912	12	103240705	103240705
Phenylketonuria	PAH	935GtoT | G312V	N/A	12	103240707	103240707
Phenylketonuria	PAH	935delG | 935del1 | 935del	N/A	12	103240707	103240707
Phenylketonuria	PAH	932TtoC | L311P	rs62642936	12	103240710	103240710
Phenylketonuria	PAH	929CtoA | S310Y	rs62642913	12	103240713	103240713
Phenylketonuria	PAH	929CtoT | S310F	rs62642913	12	103240713	103240713
Phenylketonuria	PAH	926CtoA | A309D	rs62642935	12	103240716	103240716
Hyperphenylalaninaemia	PAH	926CtoT | A309V	rs62642935	12	103240716	103240716
Phenylketonuria	PAH	922CtoG | L308V	rs62642095	12	103240720	103240720
Hyperphenylalaninaemia	PAH	922CtoT | L308F	rs62642095	12	103240720	103240720
Phenylketonuria	PAH	920delG | 920del1 | 920del	N/A	12	103240722	103240722
Phenylketonuria	PAH	916AtoG | I306V	rs62642934	12	103240726	103240726
Phenylketonuria	PAH	916delA | 916del1 | 916del	N/A	12	103240726	103240726
Phenylketonuria	PAH	913minus2AtoG	N/A	12	103240731	103240731
Phenylketonuria	PAH	913minus5TtoG	rs62507264	12	103240734	103240734
Phenylketonuria	PAH	913minus7AtoG	rs62517165	12	103240736	103240736
Phenylketonuria	PAH	802TtoC | Y268H	rs62507263	12	103246633	103246633
Phenylketonuria	PAH	801GtoC | Q267H	rs62508644	12	103246634	103246634
Phenylketonuria	PAH	800AtoT | Q267L	N/A	12	103246635	103246635
Phenylketonuria	PAH	799CtoG | Q267E	rs62508645	12	103246636	103246636
Phenylketonuria	PAH	799CtoT | Q267X	N/A	12	103246636	103246636
Phenylketonuria	PAH	796_797delACinsGA | 796_797del2ins2 | 796_797delins	N/A	12	103246638	103246639
Phenylketonuria	PAH	796AtoC | T266P	N/A	12	103246639	103246639
Phenylketonuria	PAH	794GtoA | C265Y	rs62507335	12	103246641	103246641
Phenylketonuria	PAH	793TtoG | C265G	rs62517181	12	103246642	103246642

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Phenylketonuria	PAH	791AtoT | H264L	rs62514949	12	103246644	103246644
Phenylketonuria	PAH	789CtoG | F263L	rs62642944	12	103246646	103246646
Phenylketonuria	PAH	782GtoA | R261Q	rs5030849	12	103246653	103246653
Phenylketonuria	PAH	782GtoC | R261P	N/A	12	103246653	103246653
Phenylketonuria	PAH	781CtoT | R261X	rs5030850	12	103246654	103246654
Phenylketonuria	PAH	776CtoT | A259V	rs28934900	12	103246659	103246659
Phenylketonuria	PAH	775GtoA | A259T	rs62642932	12	103246660	103246660
Phenylketonuria	PAH	773TtoC | L258P	N/A	12	103246662	103246662
Phenylketonuria	PAH	770GtoA | G257D	N/A	12	103246665	103246665
Phenylketonuria	PAH	770GtoT | G257V	rs62642908	12	103246665	103246665
Porokeratosis, disseminated superficial actinic	SSH1	188GtoA | S63N	N/A	12	109217060	109217060
Potential protein deficiency	TRPV4	2198GtoA | W733X	N/A	12	110226215	110226215
Noonan syndrome	PTPN11	5CtoT | T2I	N/A	12	112856920	112856920
Holt-Oram syndrome	TBX5	1024delT | 1024del1 | 1024del	N/A	12	114793870	114793870
Acyl-CoA-dehydrogenase deficiency	ACADS	2TtoC | M1T	N/A	12	121163690	121163690
Diabetes, MODY3	HNF1A	217GtoT | E73X	N/A	12	121416788	121416788
Diabetes, MODY3	HNF1A	225CtoA | D75E	N/A	12	121416796	121416796
Diabetes, MODY3	HNF1A	245CtoT | T82M	N/A	12	121416816	121416816
Diabetes, MODY3	HNF1A	259AtoT | K87X	N/A	12	121416830	121416830
Diabetes, MODY3	HNF1A	282dupT | 282dup1 | 282dup	N/A	12	121416853	121416854
Serum C-peptide and insulin response	HNF1A	293CtoT | A98V	rs1800574	12	121416864	121416864
Diabetes, MODY3	HNF1A	572GtoA | G191D	N/A	12	121431368	121431368
Diabetes, MODY3	HNF1A	1340CtoT | P447L	rs137853236	12	121435307	121435307
Diabetes, MODY	HNF1A	1370CtoT | T457I	N/A	12	121435337	121435337
Diabetes, MODY3	HNF1A	1369_1383dupACCACCCTGCAGCCC | 1369_1383dup15 | 1369_1383dup	N/A	12	121435350	121435351
Diabetes, MODY3	HNF1A	1387CtoT | Q463X	N/A	12	121435354	121435354
Diabetes, MODY3	HNF1A	1394CtoT | S465F	N/A	12	121435361	121435361
Diabetes, MODY	HNF1A	1396CtoT | Q466X	N/A	12	121435363	121435363
Diabetes, MODY3	HNF1A	1400CtoT | P467L	N/A	12	121435367	121435367
Diabetes, MODY3	HNF1A	1417CtoT | Q473X	N/A	12	121435384	121435384
Diabetes, MODY	HNF1A	1420CtoT | Q474X	N/A	12	121435387	121435387
Diabetes, MODY3	HNF1A	1421dupA | 1421dup1 | 1421dup	N/A	12	121435388	121435389
Diabetes, type 2, early-onset	HNF1A	1424CtoT | P475L	N/A	12	121435391	121435391
Altered function	P2RX7	1703TtoA | I568N	rs1653624	12	121622520	121622520
Spastic ataxia, Charlevoix-Saguenay	SACS	216delT | 216del1 | 216del	N/A	13	23945260	23945260
Bare lymphocyte syndrome, complementation	RFXAP	368delG | 368del1 | 368del	N/A	13	37393862	37393862
Bare lymphocyte syndrome, complementation	RFXAP	392dupG | 392dup1 | 392dup	N/A	13	37393886	37393887
HHH syndrome	SLC25A15	377CtoG | P126R	N/A	13	41379316	41379316
HHH syndrome	SLC25A15	446delG | 446del1 | 446del	N/A	13	41379385	41379385
Wilson disease	ATP7B	2121plus3AtoG	N/A	13	52534281	52534281
Wilson disease	ATP7B	2121plus3AtoT	N/A	13	52534281	52534281
Wilson disease	ATP7B	2120AtoG | Q707R	N/A	13	52534285	52534285
Wilson disease	ATP7B	2116_2117delGT | 2116_2117del2 | 2116_2117del	N/A	13	52534288	52534289
Wilson disease	ATP7B	2108GtoA | C703Y	N/A	13	52534297	52534297
Wilson disease	ATP7B	2101_2102delAT | 2101_2102del2 | 2101_2102del	N/A	13	52534303	52534304
Wilson disease	ATP7B	2097_2099delCTT | 2097_2099del3 | 2097_2099del	N/A	13	52534306	52534308
Wilson disease	ATP7B	2078CtoA | S693Y	N/A	13	52534327	52534327
Wilson disease	ATP7B	2078CtoG | S693C	N/A	13	52534327	52534327
Wilson disease	ATP7B	2077TtoC | S693P	N/A	13	52534328	52534328
Wilson disease	ATP7B	2071GtoA | G691R	rs121908001	13	52534334	52534334
Wilson disease	ATP7B	2069CtoT | P690L	N/A	13	52534336	52534336
Wilson disease	ATP7B	2038CtoT | Q680X	N/A	13	52534367	52534367
Wilson disease	ATP7B	2035delC | 2035del1 | 2035del	N/A	13	52534370	52534370
Wilson disease	ATP7B	2027AtoT | N676I	N/A	13	52534378	52534378
Wilson disease	ATP7B	2009_2015delATATGCT | 2009_2015del7 | 2009_2015del	N/A	13	52534390	52534396
Wilson disease	ATP7B	2008_2013delTATATG | 2008_2013del6 | 2008_2013del	N/A	13	52534392	52534397
Wilson disease	ATP7B	2004_2006delGAT | 2004_2006del3 | 2004_2006del	N/A	13	52534399	52534401
Wilson disease	ATP7B	2000TtoA | L667X	N/A	13	52534405	52534405
Neuronal ceroid lipofuscinosis, late infantile	CLN5	223TtoC | W75R	N/A	13	77566309	77566309
Neuronal ceroid lipofuscinosis, late infantile	CLN5	225GtoA | W75X	rs104894385	13	77566311	77566311
Neuronal ceroid lipofuscinosis, Finnish variant	CLN5	291dupC | 291dup1 | 291dup	N/A	13	77566377	77566378
Impaired taurocholate transport	SLC10A2	431GtoA | C144Y	rs143238545	13	103710679	103710679
Axenfeld-Rieger anomaly	COL4A1	2159GtoA | G720D	rs113994108	13	110833673	110833673
Small vessel disease	COL4A1	1685GtoA | G562E	rs121912857	13	110839528	110839528
Intracerebral haemorrhage, late-onset	COL4A1	1612CtoG | R538G	N/A	13	110839601	110839601
Haemorrhagic stroke	COL4A2	3368AtoG | E1123G	N/A	13	111143601	111143601
Factor VII deficiency	F7	247TtoC | S83P	N/A	13	113765120	113765120
Factor VII deficiency	F7	250_252delTTC | 250_252del3 | 250_252del	N/A	13	113765123	113765125
Factor VII deficiency	F7	253GtoA | E85K	rs121964935	13	113765126	113765126
Factor VII deficiency	F7	291plus5GtoA	N/A	13	113765169	113765169
Factor VII deficiency	F7	291plus5GtoT	N/A	13	113765169	113765169
Factor X deficiency	F10	71minus1GtoC	N/A	13	113783765	113783765
Factor X deficiency	F10	290AtoC | N97T	N/A	13	113793704	113793704
Factor X deficiency	F10	295GtoC | G99R	N/A	13	113793709	113793709
Factor X deficiency	F10	303TtoA | C101X	N/A	13	113793717	113793717
Factor X deficiency	F10	307GtoC | D103H	N/A	13	113793721	113793721
Factor X deficiency	F10	320AtoC | E107A	N/A	13	113793734	113793734
Factor X deficiency	F10	353GtoA | G118D	N/A	13	113793767	113793767
Factor X deficiency	F10	362GtoA | C121Y	N/A	13	113793776	113793776
Leber congenital amaurosis	RPGRIP1	1738AtoG | R580G	N/A	14	21790139	21790139
Lysinuric protein intolerance	SLC7A7	1005_1008delCTTT | 1005_1008del4 | 1005_1008del	N/A	14	23244740	23244743
Cardiomyopathy, hypertrophic	MYH7	3300_3311delCGGCAGCCAGCT | 3300_3311del12 | 3300_3311del	N/A	14	23890192	23890203
Cardiomyopathy, hypertrophic	MYH7	3301GtoA | G1101S	N/A	14	23890202	23890202
Cardiomyopathy, dilated	MYH7	3286GtoT | D1096Y	rs45478699	14	23890217	23890217
Cardiomyopathy, dilated	MYH7	1573GtoA | E525K	N/A	14	23897714	23897714
Ichthyosis, bathing suit	TGM1	919CtoG | R307G	rs121918731	14	24728975	24728975
Ichthyosis, lamellar	TGM1	919CtoT | R307W	rs121918731	14	24728975	24728975
Muscular dystrophy, Emery-Dreifuss	SYNE2	18632CtoT | T6211M	rs36215895	14	64676751	64676751
Retinal dystrophy	RDH12	2TtoC | M1T	N/A	14	68189361	68189361
Retinal dystrophy	RDH12	57_59delTCC | 57_59del3 | 57_59del	N/A	14	68189416	68189418
Leber congenital amaurosis	RDH12	63_66delCATC | 63_66del4 | 63_66del	N/A	14	68189422	68189425
Leber congenital amaurosis	RDH12	295CtoA | L99I	rs28940315	14	68191923	68191923
Retinal pigmentosa	RDH12	778delG | 778del1 | 778del	N/A	14	68196027	68196027
Leber congenital amaurosis	RDH12	784delG | 784del1 | 784del	N/A	14	68196033	68196033
Spastic paraplegia	ZFYVE26	5485minus1GtoA	N/A	14	68236448	68236448

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Spastic paraplegia with thin corpus callosum	ZFYVE26	5422CtoT | Q1808X	N/A	14	68238826	68238826
Spastic paraplegia with thin corpus callosum	ZFYVE26	5036delT | 5036del1 | 5036del	N/A	14	68242762	68242762
Hearing loss	ESRRB	913TtoC | Y305H	N/A	14	76957915	76957915
Hearing loss	ESRRB	959TtoC | L320P	N/A	14	76957961	76957961
Muscular dystrophy, congenital	POMT2	1997AtoG | Y666C	N/A	14	77745107	77745107
Muscular dystrophy, congenital	POMT2	1975CtoT | R659W	N/A	14	77745129	77745129
Muscular dystrophy, congenital	POMT2	1941GtoA | W647X	N/A	14	77745163	77745163
Walker-Warburg syndrome	POMT2	1912CtoT | R638X	rs119463989	14	77745192	77745192
Muscular dystrophy, congenital	POMT2	527CtoT | T176I	N/A	14	77771055	77771055
Altered enzyme activity	GSTZ1	124GtoA | G42R	rs7972	14	77793237	77793237
Hypothyroidism	TSHR	79CtoA | P27T	N/A	14	81422103	81422103
Hypothyroidism	TSHR	93CtoA | C31X	N/A	14	81422117	81422117
Hypothyroidism	TSHR	100GtoA | E34K	rs45499704	14	81422124	81422124
Thyroid-stimulating hormone resistance	TSHR	122GtoC | C41S	rs121908869	14	81422146	81422146
Hyperthyroidism	TSHR	154CtoA | P52T	rs2234919	14	81422178	81422178
Krabbe disease	GALC	967GtoA | G323R	N/A	14	88431915	88431915
Krabbe disease	GALC	956AtoG | Y319C	N/A	14	88431926	88431926
Krabbe disease	GALC	955delT | 955del1 | 955del	N/A	14	88431927	88431927
Krabbe disease	GALC	953CtoG | P318R	N/A	14	88431929	88431929
Krabbe disease	GALC	952CtoG | P318A	N/A	14	88431930	88431930
Krabbe disease	GALC	941AtoG | Y314C	N/A	14	88431941	88431941
Krabbe disease, association with	GALC	913AtoG | I305V	N/A	14	88431969	88431969
Krabbe disease	GALC	68CtoA | S23X	N/A	14	88459441	88459441
Multinodular goiter	DICER1	4050+1delG | 4050+1del1 | 4050+1del	N/A	14	95569682	95569682
Hypertension, association with	BDKRB2	NULL	rs1799722	14	96671139	96671139
TRAF3 deficiency	TRAF3	352CtoT | R118W	N/A	14	103342015	103342015
Megaloblastic anaemia 1	AMN	14delG | 14del1 | 14del	N/A	14	103389039	103389039
Megaloblastic anaemia 1	AMN	43plus1GtoT	N/A	14	103389069	103389069
Head and neck cancer, increased risk	XRCC3	281GtoA | R94H	rs3212057	14	104173465	104173465
Albinism, oculocutaneous II	OCA2	1327GtoA | V443I	rs121918166	15	28230247	28230247
Albinism, oculocutaneous II	OCA2	1290TtoA | C430X	N/A	15	28230284	28230284
Albinism, oculocutaneous II	OCA2	1275_1277delGAT | 1275_1277del3 | 1275_1277del	N/A	15	28230297	28230299
Darker eye colour, association with	OCA2	1256GtoA | R419Q	rs1800407	15	28230318	28230318
Albinism, oculocutaneous II	OCA2	1255CtoT | R419W	rs143218168	15	28230319	28230319
Albinism, ocular	OCA2	1243TtoC | Y415H	N/A	15	28230331	28230331
Albinism, oculocutaneous	OCA2	1045minus15TtoG	N/A	15	28235808	28235808
Isovaleric acidaemia	IVD	244minus1GtoC	N/A	15	40700137	40700137
Isovaleric acidaemia	IVD	271GtoT | G91C	N/A	15	40700165	40700165
Isovaleric acidaemia	IVD	290CtoG | A97G	N/A	15	40700184	40700184
Isovaleric acidaemia	IVD	295plus5GtoT	N/A	15	40700194	40700194
Potential protein deficiency	C15orf62	527AtoT | X176L	N/A	15	41063220	41063220
Muscular dystrophy, limb girdle	CAPN3	1030minus1GtoA	N/A	15	42686453	42686453
Muscular dystrophy, limb girdle	CAPN3	1043delG | 1043del1 | 1043del	N/A	15	42686467	42686467
Muscular dystrophy, limb girdle	CAPN3	1058TtoC | L353P	N/A	15	42686482	42686482
Muscular dystrophy, limb girdle	CAPN3	1061TtoC | V354A	N/A	15	42686485	42686485
Muscular dystrophy, limb girdle	CAPN3	1061TtoG | V354G	N/A	15	42686485	42686485
Muscular dystrophy, limb girdle	CAPN3	1063CtoG | R355G	N/A	15	42686487	42686487
Muscular dystrophy, limb girdle	CAPN3	1063CtoT | R355W	N/A	15	42686487	42686487
Muscular dystrophy, limb girdle	CAPN3	1069CtoT | R357W	N/A	15	42686493	42686493
Muscular dystrophy, limb girdle	CAPN3	1070GtoA | R357Q	N/A	15	42686494	42686494
Muscular dystrophy, limb girdle	CAPN3	1076CtoT | P359L	N/A	15	42686500	42686500
Muscular dystrophy, limb girdle	CAPN3	1079GtoA | W360X	N/A	15	42686503	42686503
Muscular dystrophy, limb girdle	CAPN3	1080GtoC | W360C	N/A	15	42686504	42686504
Muscular dystrophy, limb girdle	CAPN3	1099GtoA | G367S	N/A	15	42686523	42686523
Muscular dystrophy, limb girdle	CAPN3	1106GtoA | W369X	N/A	15	42686530	42686530
Congenital dysorythropoietic anaemia type 1	CDAN1	2992CtoT | R998X	N/A	15	43019923	43019923
Johanson-Blizzard syndrome	UBR1	4188CtoA | C1396X	N/A	15	43270108	43270108
Johanson-Blizzard syndrome	UBR1	2254plus2TtoC	N/A	15	43324764	43324764
Spastic paraplegia, autosomal recessive	SPG11	4868delT | 4868del1 | 4868del	N/A	15	44881488	44881488
Spastic paraplegia with thin corpus callosum	SPG11	4846CtoT | Q1616X	N/A	15	44881510	44881510
Spastic paraplegia with thin corpus callosum	SPG11	4790GtoA | W1597X	N/A	15	44881566	44881566
Hypercatabolic hypoproteinaemia, familial	B2M	31GtoC | A11P	rs104894481	15	45003775	45003775
Hypothyroidism	DUOX2	602dupG | 602dup1 | 602dup	N/A	15	45403694	45403695
Marfan syndrome	FBN1	7454minus2AtoT	N/A	15	48714267	48714267
Marfan syndrome	FBN1	6031TtoG | C2011G	N/A	15	48736744	48736744
Marfan syndrome	FBN1	6018delT | 6018del1 | 6018del	N/A	15	48736757	48736757
Marfan syndrome	FBN1	2623TtoC | C875R	N/A	15	48787374	48787374
Marfan syndrome	FBN1	2057CtoA | A686D	N/A	15	48796040	48796040
Marfan syndrome	FBN1	2056GtoA | A686T	N/A	15	48796041	48796041
Marfan syndrome	FBN1	2055CtoG | C685W	N/A	15	48796042	48796042
Marfan syndrome	FBN1	2054GtoA | C685Y	N/A	15	48796043	48796043
Marfan syndrome	FBN1	2051GtoA | C684Y	N/A	15	48796046	48796046
Fibrillinopathy	FBN1	2050TtoC | C684R	N/A	15	48796047	48796047
Marfan syndrome	FBN1	2048GtoA | C683Y	N/A	15	48796049	48796049
Marfan syndrome	FBN1	2047TtoC | C683R	N/A	15	48796050	48796050
Marfan syndrome	FBN1	2042CtoA | S681Y	N/A	15	48796055	48796055
Marfan syndrome	FBN1	2023_2026delTTTG | 2023_2026del4 | 2023_2026del	N/A	15	48796071	48796074
Marfan syndrome	FBN1	1995CtoG | Y665X	N/A	15	48796102	48796102
Marfan syndrome	FBN1	1982GtoA | C661Y	N/A	15	48796115	48796115
Marfan syndrome	FBN1	1981dupT | 1981dup1 | 1981dup	N/A	15	48796115	48796116
Marfan syndrome	FBN1	1981TtoC | C661R	N/A	15	48796116	48796116
Marfan syndrome	FBN1	1981TtoG | C661G	N/A	15	48796116	48796116
Marfan syndrome	FBN1	1971dupG | 1971dup1 | 1971dup	N/A	15	48796125	48796126
Marfan syndrome	FBN1	1969_1970delAT | 1969_1970del2 | 1969_1970del	N/A	15	48796127	48796128
Marfan syndrome	FBN1	1961minus3TtoG	N/A	15	48796139	48796139
Cardiomyopathy, non-compaction, left ventric	TPM1	109AtoG | K37E	N/A	15	63335137	63335137
Osteogenesis imperfecta	PPIB	120delC | 120del1 | 120del	rs137853865	15	64455066	64455066
Osteogenesis imperfecta, recessive	PPIB	2TtoG | M1R	N/A	15	64455184	64455184
Neuronal ceroid lipofuscinosis, late infantile	CLN6	663CtoG | Y221X	rs104894486	15	68501977	68501977
Neuronal ceroid lipofuscinosis, late infantile	CLN6	662AtoC | Y221S	N/A	15	68501978	68501978
Neuronal ceroid lipofuscinosis, late infantile	CLN6	662AtoG | Y221C	N/A	15	68501978	68501978
Neuronal ceroid lipofuscinosis, late infantile	CLN6	557TtoC | F186S	N/A	15	68502083	68502083
Neuronal ceroid lipofuscinosis, late infantile	CLN6	552dupC | 552dup1 | 552dup	N/A	15	68502087	68502088
Tay-Sachs disease	HEXA	540CtoG | Y180X	rs121907969	15	72645439	72645439
Gangliosidosis GM2	HEXA	538TtoC | Y180H	rs28941771	15	72645441	72645441

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Tay-Sachs disease	HEXA	536AtoG | H179R	N/A	15	72645443	72645443
Tay-Sachs disease	HEXA	535CtoT | H179Y	N/A	15	72645444	72645444
Tay-Sachs disease	HEXA	533GtoA | R178H	N/A	15	72645446	72645446
Tay-Sachs disease	HEXA	533GtoT | R178L	rs28941770	15	72645446	72645446
Tay-Sachs disease	HEXA	532CtoT | R178C	rs121907953	15	72645447	72645447
Tay-Sachs disease	HEXA	509GtoA | R170Q	rs121907957	15	72645470	72645470
Tay-Sachs disease	HEXA	508CtoT | R170W	rs121907972	15	72645471	72645471
Tay-Sachs disease	HEXA	498delC | 498del1 | 498del	N/A	15	72645481	72645481
Tay-Sachs disease	HEXA	496_498delCGCinsGG | 496_498del3ins2 | 496_498delins	N/A	15	72645481	72645483
Tay-Sachs disease	HEXA	496CtoG | R166G	N/A	15	72645483	72645483
Tay-Sachs disease	HEXA	496delC | 496del1 | 496del	N/A	15	72645483	72645483
Tay-Sachs disease	HEXA	477_478delTG | 477_478del2 | 477_478del	N/A	15	72645501	72645502
Tay-Sachs disease	HEXA	459plus5GtoA	N/A	15	72646027	72646027
Tay-Sachs disease	HEXA	459+2dupT | 459+2dup1 | 459+2dup	N/A	15	72646029	72646030
Microphthalmia, syndromic 9	STRA6	859CtoT | Q287X	N/A	15	74483514	74483514
XY sex reversal and adrenal insufficiency	CYP11A1	809_814dupGGGACG | 809_814dup6 | 809_814dup	N/A	15	74636144	74636145
P450scc deficiency	CYP11A1	806CtoT | A269V	N/A	15	74636153	74636153
Congenital disorder of glycosylation 1b	MPI	391GtoA | D131N	N/A	15	75185047	75185047
Congenital disorder of glycosylation 1b	MPI	413TtoC | M138T	rs104894495	15	75185069	75185069
Congenital disorder of glycosylation 1b	MPI	419TtoC | I140T	N/A	15	75185075	75185075
Electron transfer flavoprotein deficiency	ETFA	73delA | 73del1 | 73del	N/A	15	76588045	76588045
Electron transfer flavoprotein deficiency	ETFA	52CtoT | R18X	N/A	15	76588066	76588066
PAPA syndrome	PSTPIP1	688GtoA | A230T	rs28939381	15	77323566	77323566
Pyoderma gangrenosum	PSTPIP1	773GtoC | G258A	rs34240327	15	77324670	77324670
PAPA syndrome	PSTPIP1	796GtoA | D266N	rs104895418	15	77324693	77324693
Altered function	CHRNB4	1399AtoG | M467V	rs61737502	15	78917573	78917573
Tyrosinaemia 1	FAH	1141AtoG | R381G	rs121965077	15	80473462	80473462
Deafness	KIAA1199	2347CtoT | H783Y	N/A	15	81218023	81218023
Alpers syndrome	POLG	2143CtoT | Q715X	N/A	15	89867060	89867060
Acrocallosal syndrome	KIF7	3001CtoT | Q1001X	N/A	15	90174836	90174836
Bardet-Biedl syndrome	KIF7	2276TtoC | L759P	N/A	15	90185552	90185552
Intellectual disability	KIF7	2272GtoA | E758K	N/A	15	90185556	90185556
Partial lipodystrophy, dyslipidemia & insulin-re	PLIN1	1210minus1GtoT	N/A	15	90209174	90209174
Brachydactyly	CHSY1	96delC | 96del1 | 96del	N/A	15	101791566	101791566
Temtamy preaxial brachydactyly syndrome	CHSY1	14delG | 14del1 | 14del	N/A	15	101791648	101791648
Stuttering	GNPTG	15_23dupGGCGCGGCT | 15_23dup9 | 15_23dup	N/A	16	1401989	1401990
Osteopetrosis, autosomal dominant, Type 2	CLCN7	2062_2064delCTA | 2062_2064del3 | 2062_2064del	N/A	16	1497665	1497667
Osteopetrosis, autosomal dominant, Type 2	CLCN7	954CtoG | F318L	N/A	16	1505759	1505759
Osteopetrosis, autosomal dominant, Type 2	CLCN7	947CtoG | A316G	N/A	16	1505766	1505766
Osteopetrosis, autosomal dominant, Type 2	CLCN7	937GtoA | E313K	N/A	16	1505776	1505776
Infantile malignant osteopetrosis	CLCN7	889GtoA | V297M	N/A	16	1506141	1506141
Osteopetrosis, autosomal dominant, Type 2	CLCN7	869CtoT | S290F	N/A	16	1506161	1506161
Osteopetrosis, autosomal dominant, Type 2	CLCN7	857GtoA | R286Q	N/A	16	1506173	1506173
Osteopetrosis, autosomal dominant, Type 2	CLCN7	856CtoT | R286W	N/A	16	1506174	1506174
Tuberous sclerosis	TSC2	292CtoT | R98W	N/A	16	2103409	2103409
Tuberous sclerosis	TSC2	299_300dupCG | 299_300dup2 | 299_300dup	N/A	16	2103417	2103418
Tuberous sclerosis	TSC2	307_315dupGCTCTGCTG | 307_315dup9 | 307_315dup	N/A	16	2103432	2103433
Tuberous sclerosis	TSC2	330delGinsAT | 330del1ins2 | 330delins	rs137854108	16	2103447	2103447
Tuberous sclerosis	TSC2	1258minus3CtoG	rs45517161	16	2112495	2112495
Tuberous sclerosis	TSC2	1258minus2AtoC	rs45517160	16	2112496	2112496
Tuberous sclerosis	TSC2	1258minus1GtoA	N/A	16	2112497	2112497
Tuberous sclerosis	TSC2	1266_1267delCC | 1266_1267del2 | 1266_1267del	rs137854238	16	2112506	2112507
Tuberous sclerosis	TSC2	1283_1285delCCT | 1283_1285del3 | 1283_1285del	N/A	16	2112523	2112525
Tuberous sclerosis	TSC2	1287TtoG | Y429X	N/A	16	2112527	2112527
Tuberous sclerosis	TSC2	1322GtoA | W441X	rs45515894	16	2112562	2112562
Tuberous sclerosis	TSC2	1327CtoT | Q443X	N/A	16	2112567	2112567
Tuberous sclerosis	TSC2	1336CtoT | Q446X	rs45517163	16	2112576	2112576
Tuberous sclerosis	TSC2	1347GtoC | M449I	rs45443091	16	2112587	2112587
Tuberous sclerosis	TSC2	1348GtoT | E450X	rs45491095	16	2112588	2112588
Tuberous sclerosis	TSC2	1352_1353delGA | 1352_1353del2 | 1352_1353del	N/A	16	2112592	2112593
Tuberous sclerosis	TSC2	1361plus2TtoG	N/A	16	2112603	2112603
Tuberous sclerosis	TSC2	2024_2042delCAGGCCCCGCCGTGCGGCT | 2024_2042del19 | 2024_2042del	rs137854309	16	2121862	2121880
Tuberous sclerosis	TSC2	2399dupG | 2399dup1 | 2399dup	N/A	16	2124244	2124245
Tuberous sclerosis	TSC2	2404dupA | 2404dup1 | 2404dup	N/A	16	2124249	2124250
Tuberous sclerosis	TSC2	2410TtoC | C804R	rs137853995	16	2124255	2124255
Tuberous sclerosis	TSC2	2498_2502delTCTCA | 2498_2502del5 | 2498_2502del	rs137854286	16	2124343	2124347
Tuberous sclerosis	TSC2	2525delC | 2525del1 | 2525del	N/A	16	2124370	2124370
Tuberous sclerosis	TSC2	2530delC | 2530del1 | 2530del	rs137854372	16	2124375	2124375
Tuberous sclerosis	TSC2	2539delC | 2539del1 | 2539del	rs137854140	16	2124384	2124384
Tuberous sclerosis	TSC2	2539dupC | 2539dup1 | 2539dup	N/A	16	2124384	2124385
Tuberous sclerosis	TSC2	2540TtoC | L847P	rs45517240	16	2124385	2124385
Tuberous sclerosis	TSC2	3532CtoT | Q1178X	rs45517297	16	2130300	2130300
Tuberous sclerosis	TSC2	3535GtoT | E1179X	rs45483391	16	2130303	2130303
Tuberous sclerosis	TSC2	4493+1delG | 4493+1del1 | 4493+1del	N/A	16	2134717	2134717
Polycystic kidney disease 1	PKD1	11799delC | 11799del1 | 11799del	N/A	16	2141089	2141089
Polycystic kidney disease 1	PKD1	11766GtoA | W3922X	N/A	16	2141122	2141122
Polycystic kidney disease 1	PKD1	11752GtoT | E3918X	N/A	16	2141136	2141136
Polycystic kidney disease 1	PKD1	11621delC | 11621del1 | 11621del	N/A	16	2141515	2141515
Polycystic kidney disease 1	PKD1	11614GtoT | E3872X	N/A	16	2141522	2141522
Polycystic kidney disease 1	PKD1	11555TtoC | L3852P	N/A	16	2141581	2141581
Polycystic kidney disease, autosomal dominan	PKD1	11554delC | 11554del1 | 11554del	N/A	16	2141582	2141582
Polycystic kidney disease 1	PKD1	11538minus2AtoG	N/A	16	2141600	2141600
Polycystic kidney disease 1	PKD1	11156GtoA | R3719Q	N/A	16	2142955	2142955
Chronic lung disease	ABCA3	3905GtoA | G1302E	N/A	16	2333317	2333317
Chronic lung disease	ABCA3	3902CtoT | P1301L	N/A	16	2333320	2333320
Fatal surfactant deficiency	ABCA3	2741AtoG | K914R	N/A	16	2338290	2338290
Neonatal respiratory distress / interstitial lung	ABCA3	2125CtoT | R709W	rs148671332	16	2347468	2347468
Lung disease, interstitial	ABCA3	2068GtoA | E690K	N/A	16	2347525	2347525
Fatal surfactant deficiency	ABCA3	464GtoA | R155Q	N/A	16	2373673	2373673
Mediterranean fever, familial	MEFV	265GtoA | A89T	rs104895124	16	3306323	3306323
Mediterranean fever, familial	MEFV	250GtoA | E84K	rs150819742	16	3306338	3306338
Breast cancer	SLX4	2098GtoA | G700R	N/A	16	3644516	3644516
Rubinstein-Taybi syndrome	CREBBP	5793dupC | 5793dup1 | 5793dup	N/A	16	3779254	3779255
Rubinstein-Taybi syndrome	CREBBP	1984CtoT | Q662X	N/A	16	3828141	3828141
Congenital disorder of glycosylation 1a	PMM2	686AtoC | Y229S	N/A	16	8941627	8941627

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Charcot-Marie-Tooth disease 1c	LITAF	430GtoA | V144M	rs121908615	16	11643549	11643549
Charcot-Marie-Tooth disease 1c	LITAF	403CtoA | P135T	N/A	16	11643576	11643576
Charcot-Marie-Tooth disease 1c	LITAF	403CtoT | P135S	N/A	16	11643576	11643576
Lung cancer, susceptibility to	ERCC4	8CtoT | S3L	rs148904556	16	14014030	14014030
Lung cancer, susceptibility to	ERCC4	16CtoT | P6S	rs61760160	16	14014038	14014038
Pseudoxanthoma elasticum, autosomal recess	ABCC6	3803GtoA | R1268Q	rs2238472	16	16251599	16251599
Pseudoxanthoma elasticum, autosomal recess	ABCC6	3787GtoA | G1263R	N/A	16	16251615	16251615
Pseudoxanthoma elasticum	ABCC6	3735GtoA	N/A	16	16253339	16253339
Pseudoxanthoma elasticum	ABCC6	3735GtoT	N/A	16	16253339	16253339
Pseudoxanthoma elasticum	ABCC6	3723GtoC | W1241C	rs72657701	16	16253351	16253351
Pseudoxanthoma elasticum, autosomal recess	ABCC6	3722GtoA | W1241X	rs72653748	16	16253352	16253352
Pseudoxanthoma elasticum	ABCC6	3717TtoG | Y1239X	rs72653747	16	16253357	16253357
Pseudoxanthoma elasticum	ABCC6	3715TtoC | Y1239H	rs63749992	16	16253359	16253359
Pseudoxanthoma elasticum, autosomal recess	ABCC6	3712GtoC | D1238H	rs63749796	16	16253362	16253362
Pseudoxanthoma elasticum, autosomal domin	ABCC6	3709CtoT | Q1237X	rs72653746	16	16253365	16253365
Pseudoxanthoma elasticum	ABCC6	3703CtoT | R1235W	rs63750402	16	16253371	16253371
Pseudoxanthoma elasticum, autosomal recess	ABCC6	3676CtoA | L1226I	rs63750125	16	16253398	16253398
Pseudoxanthoma elasticum, autosomal recess	ABCC6	3668GtoA | W1223X	rs72653745	16	16253406	16253406
Pseudoxanthoma elasticum, autosomal domin	ABCC6	3608GtoA | G1203D	rs63750607	16	16255320	16255320
Pseudoxanthoma elasticum	ABCC6	3507minus1GtoA	rs72664210	16	16255422	16255422
Pseudoxanthoma elasticum, autosomal recess	ABCC6	3507minus3CtoA	N/A	16	16255424	16255424
Pseudoxanthoma elasticum, autosomal recess	ABCC6	3507minus3CtoT	rs41278172	16	16255424	16255424
Pseudoxanthoma elasticum, autosomal recess	ABCC6	2943GtoT | Q981H	N/A	16	16263555	16263555
Pseudoxanthoma elasticum	ABCC6	2891GtoC | R964P	rs72657691	16	16263607	16263607
Pseudoxanthoma elasticum	ABCC6	2855TtoG | F952C	rs72657690	16	16263643	16263643
Pseudoxanthoma elasticum	ABCC6	2836_2851delCTCTGCCTCTACGCAC | 2836_2851del16 | 2836_2851del	rs74315152	16	16263647	16263662
Angioid streaks	ABCC6	2849CtoT | A950V	rs190557767	16	16263649	16263649
Pseudoxanthoma elasticum, autosomal recess	ABCC6	2848GtoA | A950T	rs72657689	16	16263650	16263650
Pseudoxanthoma elasticum	ABCC6	2836CtoA | L946I	rs61340537	16	16263662	16263662
Pseudoxanthoma elasticum, autosomal recess	ABCC6	2831CtoT | T944I	rs72653801	16	16263667	16263667
Pseudoxanthoma elasticum, autosomal recess	ABCC6	2820_2821insC | 2820_2821ins1 | 2820_2821ins	rs72664219	16	16263677	16263678
Pseudoxanthoma elasticum, autosomal recess	ABCC6	2814CtoG | Y938X	rs72653704	16	16263684	16263684
Pseudoxanthoma elasticum	ABCC6	2030TtoC | L677P	rs72653782	16	16276701	16276701
Pseudoxanthoma elasticum, autosomal domin	ABCC6	2018TtoC | L673P	rs67470842	16	16276713	16276713
Pseudoxanthoma elasticum, autosomal recess	ABCC6	1999delG | 1999del1 | 1999del	rs72664228	16	16276732	16276732
Pseudoxanthoma elasticum, autosomal recess	ABCC6	1997GtoT | G666V	N/A	16	16276734	16276734
Pseudoxanthoma elasticum, autosomal recess	ABCC6	1987GtoT | G663C	rs72653780	16	16276744	16276744
Pseudoxanthoma elasticum	ABCC6	1798CtoT | R600C	rs72653777	16	16281050	16281050
Pseudoxanthoma elasticum	ABCC6	1781CtoT | A594V	rs72653776	16	16281067	16281067
Pseudoxanthoma elasticum	ABCC6	1780minus29TtoA	rs72664206	16	16281097	16281097
Tubulointerstitial nephritis	UMOD	514GtoC | D172H	N/A	16	20360109	20360109
Breast cancer, male	PALB2	3539TtoC | I1180T	rs180177139	16	23614802	23614802
Fanconi anaemia, N type	PALB2	3294_3298delGACGA | 3294_3298del5 | 3294_3298del	rs180177134	16	23619237	23619241
Breast cancer	PALB2	48plus1GtoC	N/A	16	23652430	23652430
Breast cancer	PALB2	23CtoT | P8L	rs150390726	16	23652456	23652456
Breast cancer	PALB2	13CtoT | P5S	N/A	16	23652466	23652466
Diabetes, type 1, association with	IL4R	1199AtoC | E400A	rs1805011	16	27373872	27373872
Paroxysmal kinesigenic dyskinesia	PRRT2	487CtoT | Q163X	N/A	16	29824862	29824862
Cardiomyopathy, dilated	CTF1	274GtoA | A92T	rs2234933	16	30913528	30913528
3 beta-hydroxysteroid oxidoreductase deficier	HSD3B7	76_78delGTG | 76_78del3 | 76_78del	N/A	16	30997055	30997057
3 beta-hydroxysteroid oxidoreductase deficier	HSD3B7	115_116insCC | 115_116ins2 | 115_116ins	N/A	16	30997094	30997095
3 beta-hydroxysteroid oxidoreductase deficier	HSD3B7	123_124delCT | 123_124del2 | 123_124del	N/A	16	30997102	30997103
Amyotrophic lateral sclerosis	FUS	730CtoT | R244C	N/A	16	31196466	31196466
Crohn's disease, susceptibility to	NOD2	113GtoT | R38M	rs104895487	16	50733438	50733438
Brooke-Spiegler syndrome	CYLD	1096_1097delCA | 1096_1097del2 | 1096_1097del	N/A	16	50811810	50811811
Cylindromatosis	CYLD	1112CtoA | S371X	N/A	16	50811826	50811826
Cylindromatosis	CYLD	1135GtoT | E379X	N/A	16	50811849	50811849
Cylindromatosis	CYLD	1569TtoG | Y523X	N/A	16	50815207	50815207
Trichoepithelioma, multiple familial	CYLD	1628_1629delCT | 1628_1629del2 | 1628_1629del	N/A	16	50815266	50815267
Cylindromatosis	CYLD	1681_1682delTT | 1681_1682del2 | 1681_1682del	N/A	16	50815319	50815320
Trichoepithelioma, multiple familial	CYLD	1682TtoA | L561X	N/A	16	50815320	50815320
Brooke-Spiegler syndrome	CYLD	1684plus1GtoA	N/A	16	50815323	50815323
Joubert syndrome	RPGRIP1L	3295minus2AtoG	N/A	16	53656270	53656270
Gitelman syndrome	SLC12A3	1AtoT | M1L	N/A	16	56899148	56899148
Gitelman syndrome	SLC12A3	3GtoA | M1I	N/A	16	56899150	56899150
Gitelman syndrome	SLC12A3	20_21delCA | 20_21del2 | 20_21del	N/A	16	56899167	56899168
Salt-losing tubulopathy	SLC12A3	37GtoC | A13P	N/A	16	56899184	56899184
Gitelman syndrome	SLC12A3	43_44delTT | 43_44del2 | 43_44del	N/A	16	56899190	56899191
Gitelman syndrome	SLC12A3	46_47delTG | 46_47del2 | 46_47del	N/A	16	56899193	56899194
Gitelman syndrome	SLC12A3	83_84insGC | 83_84ins2 | 83_84ins	N/A	16	56899230	56899231
Gitelman syndrome	SLC12A3	104delCinsTT | 104del1ins2 | 104delins	N/A	16	56899251	56899251
Gitelman syndrome	SLC12A3	111TtoA | Y37X	N/A	16	56899258	56899258
Gitelman syndrome	SLC12A3	160CtoT | R54C	N/A	16	56899307	56899307
Gitelman syndrome	SLC12A3	174CtoA | Y58X	N/A	16	56899321	56899321
Gitelman syndrome	SLC12A3	176AtoT | N59I	N/A	16	56899323	56899323
Gitelman syndrome	SLC12A3	179CtoT | T60M	N/A	16	56899326	56899326
Gitelman syndrome	SLC12A3	184GtoA | D62N	N/A	16	56899331	56899331
Gitelman syndrome	SLC12A3	184GtoC | D62H	N/A	16	56899331	56899331
Salt-losing tubulopathy	SLC12A3	185AtoG | D62G	N/A	16	56899332	56899332
Gitelman syndrome	SLC12A3	190_192delGTG | 190_192del3 | 190_192del	N/A	16	56899337	56899339
Gitelman syndrome	SLC12A3	202GtoA | E68K	N/A	16	56899349	56899349
Gitelman syndrome	SLC12A3	205CtoA | H69N	N/A	16	56899352	56899352
Gitelman syndrome	SLC12A3	237_238dupCC | 237_238dup2 | 237_238dup	N/A	16	56899385	56899386
Gitelman syndrome	SLC12A3	247CtoT | R83W	N/A	16	56899394	56899394
Gitelman syndrome	SLC12A3	248GtoA | R83Q	N/A	16	56899395	56899395
Gitelman syndrome	SLC12A3	265CtoG | L89V	N/A	16	56899412	56899412
Gitelman syndrome	SLC12A3	268CtoT | H90Y	N/A	16	56899415	56899415
Gitelman syndrome	SLC12A3	363GtoC | E121D	N/A	16	56901062	56901062
Gitelman syndrome	SLC12A3	433CtoT | R145C	rs148945966	16	56902212	56902212
Gitelman syndrome	SLC12A3	434GtoA | R145H	N/A	16	56902213	56902213
Gitelman syndrome	SLC12A3	437GtoT | C146F	N/A	16	56902216	56902216
Gitelman syndrome	SLC12A3	443TtoC | L148P	N/A	16	56902222	56902222
Gitelman syndrome	SLC12A3	450TtoG | I150M	N/A	16	56902229	56902229
Gitelman syndrome	SLC12A3	457GtoA | V153M	N/A	16	56902236	56902236
Gitelman syndrome	SLC12A3	460AtoT | I154F	N/A	16	56902239	56902239

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Gitelman syndrome	SLC12A3	1367delT | 1367del1 | 1367del	N/A	16	56913485	56913485
Gitelman syndrome	SLC12A3	1376CtoA | A459D	N/A	16	56913494	56913494
Gitelman syndrome	SLC12A3	1379GtoA | G460D	N/A	16	56913497	56913497
Gitelman syndrome	SLC12A3	1379delG | 1379del1 | 1379del	N/A	16	56913497	56913497
Gitelman syndrome	SLC12A3	1387GtoA | G463R	N/A	16	56913505	56913505
Gitelman syndrome	SLC12A3	1388GtoA | G463E	N/A	16	56913506	56913506
Gitelman syndrome	SLC12A3	1390GtoA | A464T	N/A	16	56913508	56913508
Gitelman syndrome	SLC12A3	1393AtoC | T465P	N/A	16	56913511	56913511
Gitelman syndrome	SLC12A3	1394CtoA | T465N	rs146392700	16	56913512	56913512
Salt-losing tubulopathy	SLC12A3	1421delT | 1421del1 | 1421del	N/A	16	56913539	56913539
Gitelman syndrome	SLC12A3	1424CtoG | S475C	N/A	16	56913542	56913542
Gitelman syndrome	SLC12A3	1432AtoG | K478E	N/A	16	56913550	56913550
Retinitis pigmentosa	CNGB1	2284CtoT | R762C	N/A	16	57949173	57949173
Retinitis pigmentosa	CNGB1	413minus1GtoA	rs189234741	16	57996515	57996515
Poikiloderma, clericuzio-type with neutropeni	C16orf57	504minus2AtoC	N/A	16	58051236	58051236
Autoimmune lymphoproliferative syndrome	TRADD	197GtoA | R66H	N/A	16	67189512	67189512
Apparent mineralocorticoid excess	HSD11B2	77_78delCA | 77_78del2 | 77_78del	N/A	16	67465228	67465229
Lecithin:cholesterol acyltransferase deficiency	LCAT	86AtoT | N29I	N/A	16	67977919	67977919
Hypotrichosis with juvenile macular dystrophy	CDH3	160plus1GtoA	N/A	16	68679654	68679654
Gastric cancer	CDH1	2TtoC | M1T	N/A	16	68771320	68771320
Gastric cancer	CDH1	3GtoA | M1I	N/A	16	68771321	68771321
Gastric cancer	CDH1	3GtoC | M1I	N/A	16	68771321	68771321
Breast cancer	CDH1	8CtoG | P3R	N/A	16	68771326	68771326
Gastric cancer	CDH1	41delT | 41del1 | 41del	N/A	16	68771359	68771359
Gastric cancer	CDH1	44dupT | 44dup1 | 44dup	N/A	16	68771362	68771363
Gastric cancer	CDH1	44_46dupTGC | 44_46dup3 | 44_46dup	N/A	16	68771364	68771365
Gastric cancer	CDH1	48plus1GtoA	N/A	16	68771367	68771367
Tyrosinaemia 2	TAT	818TtoC | L273P	N/A	16	71604676	71604676
Tyrosinaemia 2	TAT	668CtoG | S223X	rs118203915	16	71606127	71606127
Tyrosinaemia 2	TAT	658CtoT | P220S	N/A	16	71606137	71606137
Tyrosinaemia 2	TAT	355CtoT | R119W	N/A	16	71607495	71607495
Leukodystrophy with spastic paraparesis & dys	FA2H	786plus1GtoA	N/A	16	74752885	74752885
Spastic paraplegia	FA2H	159_176delGGCGGGCCAGGACATCAG | 159_176del18 | 159_176del	N/A	16	74808478	74808495
Leukodystrophy with spastic paraparesis & dys	FA2H	103GtoT | D35Y	rs121918217	16	74808551	74808551
Macular corneal dystrophy, type 1	CHST6	6_7delGCinsAA | 6_7del2ins2 | 6_7delins	N/A	16	75513720	75513721
Giant axonal neuropathy	GAN	145GtoA | A49T	N/A	16	81348863	81348863
Giant axonal neuropathy	GAN	151GtoC | A51P	N/A	16	81348869	81348869
Giant axonal neuropathy	GAN	154AtoG | S52G	N/A	16	81348872	81348872
Malonyl-CoA decarboxylase deficiency	MLYCD	119TtoC | M40T	rs28937908	16	83932868	83932868
Malonyl-CoA decarboxylase deficiency	MLYCD	475delG | 475del1 | 475del	N/A	16	83933224	83933224
Malonyl-CoA decarboxylase deficiency	MLYCD	481delC | 481del1 | 481del	N/A	16	83933230	83933230
Malonyl-CoA decarboxylase deficiency	MLYCD	482TtoC | L161P	N/A	16	83933231	83933231
Autism	KIAA0182	1196GtoA | R399Q	N/A	16	85691078	85691078
Immunodeficiency, autosomal recessive	IRF8	322AtoG | K108E	N/A	16	85942743	85942743
Chronic granulomatous disease	CYBA	408delC | 408del1 | 408del	N/A	16	88709941	88709941
Chronic granulomatous disease	CYBA	354CtoA | S118R	rs104894514	16	88712539	88712539
Chronic granulomatous disease	CYBA	339CtoA | C113X	N/A	16	88712554	88712554
Chronic granulomatous disease	CYBA	295_301delGTGCCCG | 295_301del7 | 295_301del	N/A	16	88712592	88712598
Chronic granulomatous disease	CYBA	288-3_300delCAGGCTCTCGGTGCCC | 288-3_300del16 | 288-3_300del	N/A	16	88712593	88712608
Chronic granulomatous disease	CYBA	288GtoT	N/A	16	88712605	88712605
Chronic granulomatous disease	CYBA	269GtoA | R90Q	rs104894513	16	88713181	88713181
Chronic granulomatous disease	CYBA	269GtoC | R90P	N/A	16	88713181	88713181
Chronic granulomatous disease	CYBA	268CtoG | R90G	N/A	16	88713182	88713182
Chronic granulomatous disease	CYBA	268CtoT | R90W	rs179363892	16	88713182	88713182
Chronic granulomatous disease	CYBA	261CtoA | Y87X	N/A	16	88713189	88713189
Chronic granulomatous disease	CYBA	261CtoG | Y87X	N/A	16	88713189	88713189
Chronic granulomatous disease	CYBA	246delC | 246del1 | 246del	N/A	16	88713204	88713204
Chronic granulomatous disease	CYBA	223delG | 223del1 | 223del	N/A	16	88713227	88713227
Coronary artery disease, lower risk, associatio	CYBA	214CtoT | H72Y	rs4673	16	88713236	88713236
Reduced promoter activity	CYBA	NULL	rs9932581	16	88718353	88718353
Meier-Gorlin syndrome	CDT1	196GtoA | A66T	N/A	16	88870435	88870435
Adenine phosphoribosyltransferase deficiency	APRT	182dupT | 182dup1 | 182dup	N/A	16	88877962	88877963
Adenine phosphoribosyltransferase deficiency	APRT	119GtoC | R40P	N/A	16	88878026	88878026
Adenine phosphoribosyltransferase deficiency	APRT	98TtoC | L33P	N/A	16	88878047	88878047
Adenine phosphoribosyltransferase deficiency	APRT	84CtoA | D28E	N/A	16	88878061	88878061
Adenine phosphoribosyltransferase deficiency	APRT	3GtoA | M1I	N/A	16	88878305	88878305
Adenine phosphoribosyltransferase deficiency	APRT	1AtoG | M1V	N/A	16	88878307	88878307
Mucopolysaccharidosis IVa	GALNS	631CtoT | Q211X	N/A	16	88902611	88902611
Mucopolysaccharidosis IVa	GALNS	612CtoG | N204K	rs118204435	16	88902630	88902630
Mucopolysaccharidosis IVa	GALNS	608CtoT | A203V	N/A	16	88902634	88902634
Mucopolysaccharidosis IVa	GALNS	604delG | 604del1 | 604del	N/A	16	88902638	88902638
Mucopolysaccharidosis IVa	GALNS	554AtoG | E185G	N/A	16	88904042	88904042
Mucopolysaccharidosis IVa	GALNS	536CtoA | P179H	N/A	16	88904060	88904060
Mucopolysaccharidosis IVa	GALNS	536CtoT | P179L	N/A	16	88904060	88904060
Mucopolysaccharidosis IVa	GALNS	535CtoT | P179S	N/A	16	88904061	88904061
Mucopolysaccharidosis IVa	GALNS	532_533delAT | 532_533del2 | 532_533del	N/A	16	88904063	88904064
Mucopolysaccharidosis IVa	GALNS	425AtoG | H142R	N/A	16	88904171	88904171
Mucopolysaccharidosis IVa	GALNS	423GtoT | W141C	N/A	16	88904173	88904173
Mucopolysaccharidosis IVa	GALNS	423minus1GtoA	N/A	16	88904174	88904174
Mucopolysaccharidosis IVa	GALNS	67GtoC | G23R	N/A	16	88923219	88923219
Mucopolysaccharidosis IVa	GALNS	43CtoA | L15M	N/A	16	88923243	88923243
Mucopolysaccharidosis IVa	GALNS	29GtoA | W10X	N/A	16	88923257	88923257
Mucopolysaccharidosis IVa	GALNS	1AtoG | M1V	N/A	16	88923285	88923285
Intellectual disability	CDH15	274CtoT | R92W	rs121434540	16	89246680	89246680
Fanconi anaemia	FANCA	3971CtoT | P1324L	rs182657062	16	89805925	89805925
Fanconi anaemia	FANCA	3568CtoT | Q1190X	N/A	16	89811425	89811425
Fanconi anaemia	FANCA	3558dupG | 3558dup1 | 3558dup	N/A	16	89811434	89811435
Fanconi anaemia	FANCA	3520_3522delTGG | 3520_3522del3 | 3520_3522del	N/A	16	89811471	89811473
Fanconi anaemia	FANCA	3403_3405delTTC | 3403_3405del3 | 3403_3405del	N/A	16	89813242	89813244
Fanconi anaemia	FANCA	2314CtoT | Q772X	N/A	16	89836576	89836576
Fanconi anaemia	FANCA	2303TtoC | L768P	N/A	16	89836587	89836587
Fanconi anaemia	FANCA	2172dupG | 2172dup1 | 2172dup	N/A	16	89837021	89837022
Fanconi anaemia	FANCA	2170AtoC | T724P	N/A	16	89837024	89837024
Fanconi anaemia	FANCA	2167_2169delCTG | 2167_2169del3 | 2167_2169del	N/A	16	89837025	89837027
Fanconi anaemia	FANCA	1085TtoC | L362P	N/A	16	89858475	89858475

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Slower APCE cleavage time, association with	SERPINF2	97CtoT | R33W	rs2070863	17	1648502	1648502
Canavan disease	ASPA	770CtoG | P257R	N/A	17	3402210	3402210
Amyotrophic lateral sclerosis	PFN1	353GtoT | G118V	N/A	17	4849265	4849265
Very long chain acyl-CoA dehydrogenase defic	ACADVL	520GtoA | V174M	N/A	17	7124899	7124899
Male factor infertility, association with	YBX2	239CtoA | P80Q	rs189257850	17	7197581	7197581
Infertility, protection against, association with	YBX2	187TtoC | S63P	rs8069533	17	7197633	7197633
Li-Fraumeni syndrome	TP53	772GtoA | E258K	rs121912652	17	7577509	7577509
Li-Fraumeni syndrome	TP53	770TtoA | L257Q	rs28934577	17	7577511	7577511
Li-Fraumeni syndrome	TP53	770delT | 770del1 | 770del	N/A	17	7577511	7577511
Li-Fraumeni syndrome	TP53	755TtoC | L252P	rs121912653	17	7577526	7577526
Glioma	TP53	753CtoG | I251M	N/A	17	7577528	7577528
Li-Fraumeni syndrome	TP53	751AtoC | I251L	N/A	17	7577530	7577530
Colorectal cancer	TP53	749CtoT | P250L	N/A	17	7577532	7577532
Rhabdomyosarcoma	TP53	679TtoA | S227T	N/A	17	7577602	7577602
Li-Fraumeni syndrome	TP53	673minus1GtoA	N/A	17	7577609	7577609
Li-Fraumeni syndrome	TP53	673minus2AtoG	N/A	17	7577610	7577610
Leber congenital amaurosis	GUCY2D	448TtoC | W150R	N/A	17	7906813	7906813
Leber congenital amaurosis	GUCY2D	450GtoA | W150X	N/A	17	7906815	7906815
Leber congenital amaurosis	GUCY2D	2182GtoA | D728N	N/A	17	7916489	7916489
Leber congenital amaurosis	GUCY2D	2200_2201delATinsGC | 2200_2201del2ins2 | 2200_2201delins	N/A	17	7916507	7916508
Leber congenital amaurosis	GUCY2D	2201TtoC | I734T	N/A	17	7916508	7916508
Leber congenital amaurosis	GUCY2D	2234delC | 2234del1 | 2234del	N/A	17	7916541	7916541
Leber congenital amaurosis	GUCY2D	2237AtoG | Y746C	rs61750166	17	7916544	7916544
Leber congenital amaurosis	GUCY2D	2248GtoT | E750X	rs61750167	17	7916555	7916555
Collodion membrane, self-healing	ALOX12B	1207CtoT | H403Y	N/A	17	7980376	7980376
Ichthyosis, congenital, autosomal recessive	ALOX12B	1078CtoG | Q360E	N/A	17	7980505	7980505
Ichthyosis, congenital, autosomal recessive	ALOXE3	1630CtoT | Q544X	N/A	17	8011840	8011840
Spondylocostal dysostosis	HES7	556GtoT | D186Y	N/A	17	8025011	8025011
Cytochrome c oxidase deficiency	COX10	674CtoT | P225L	rs104894556	17	14063243	14063243
Dejerine-Sottas syndrome	PMP22	299GtoA | G100E	N/A	17	15142808	15142808
Neuropathy, liability to pressure palsies	PMP22	281dupG | 281dup1 | 281dup	N/A	17	15142825	15142826
Charcot-Marie-Tooth disease 1a	PMP22	281delG | 281del1 | 281del	N/A	17	15142826	15142826
Charcot-Marie-Tooth disease 1	PMP22	277GtoC | G93R	N/A	17	15142830	15142830
Charcot-Marie-Tooth disease 1	PMP22	256CtoT | Q86X	N/A	17	15142851	15142851
Dejerine-Sottas syndrome	PMP22	251_253delTCT | 251_253del3 | 251_253del	N/A	17	15142854	15142856
Dejerine-Sottas syndrome	PMP22	239TtoC | L80P	N/A	17	15142868	15142868
Dejerine-Sottas syndrome	PMP22	239TtoG | L80R	N/A	17	15142868	15142868
Dejerine-Sottas syndrome	PMP22	238_239delCT | 238_239del2 | 238_239del	N/A	17	15142868	15142869
Charcot-Marie-Tooth disease 1a	PMP22	236CtoG | S79C	rs104894618	17	15142871	15142871
Dejerine-Sottas syndrome	PMP22	235TtoA | S79T	N/A	17	15142872	15142872
Dejerine-Sottas syndrome	PMP22	235TtoC | S79P	N/A	17	15142872	15142872
Charcot-Marie-Tooth disease 1	PMP22	233TtoC | L78P	N/A	17	15142874	15142874
Dejerine-Sottas syndrome	PMP22	227GtoT | S76I	N/A	17	15142880	15142880
Neuropathy, liability to pressure palsies	PMP22	227delG | 227del1 | 227del	N/A	17	15142880	15142880
Mitochondrial complex III deficiency	TTC19	880CtoT | Q294X	N/A	17	15907199	15907199
Immunodeficiency, common variable	TNFRSF13B	563AtoT | K188M	rs74811083	17	16843708	16843708
Immunodeficiency, common variable	TNFRSF13B	542CtoA | A181E	rs72553883	17	16843729	16843729
Immunodeficiency, common variable	TNFRSF13B	515GtoA | C172Y	N/A	17	16843756	16843756
Immunodeficiency, common variable	TNFRSF13B	512TtoG | L171R	rs143027621	17	16843759	16843759
Immunodeficiency, common variable	TNFRSF13B	171GtoC | Q57H	N/A	17	16855788	16855788
Immunodeficiency, common variable	TNFRSF13B	124CtoA | P42T	N/A	17	16855835	16855835
Birt-Hogg-DubÃ© syndrome	FLCN	1533_1536delGATG | 1533_1536del4 | 1533_1536del	N/A	17	17118301	17118304
Birt-Hogg-DubÃ© syndrome	FLCN	1533GtoA | W511X	N/A	17	17118304	17118304
Birt-Hogg-DubÃ© syndrome	FLCN	1528_1530delGAG | 1528_1530del3 | 1528_1530del	N/A	17	17118307	17118309
Birt-Hogg-DubÃ© syndrome	FLCN	1522_1524delAAG | 1522_1524del3 | 1522_1524del	N/A	17	17118313	17118315
Birt-Hogg-DubÃ© syndrome	FLCN	1523AtoG | K508R	N/A	17	17118314	17118314
Birt-Hogg-DubÃ© syndrome	FLCN	1487_1490dupCTGT | 1487_1490dup4 | 1487_1490dup	N/A	17	17118346	17118347
Birt-Hogg-DubÃ© syndrome	FLCN	1252delC | 1252del1 | 1252del	N/A	17	17119742	17119742
Birt-Hogg-DubÃ© syndrome	FLCN	1215CtoG | Y405X	N/A	17	17119779	17119779
Birt-Hogg-DubÃ© syndrome	FLCN	1183_1198delCTTCCCGTGGGCTGCG | 1183_1198del16 | 1183_1198del	N/A	17	17119796	17119811
Birt-Hogg-DubÃ© syndrome	FLCN	1177minus2AtoG	N/A	17	17119819	17119819
Birt-Hogg-DubÃ© syndrome	FLCN	1177-5_1177-3delCTC | 1177-5_1177-3del3 | 1177-5_1177-3del	N/A	17	17119820	17119822
Deafness, non-syndromic, autosomal recessive	MYO15A	3685CtoT | Q1229X	rs121908968	17	18027872	18027872
Deafness, non-syndromic, autosomal recessive	MYO15A	7982CtoA | S2661X	N/A	17	18057104	18057104
Increased promoter activity	SLC47A1	NULL	rs111427955	17	19437209	19437209
Sjoegren-Larsson syndrome	ALDH3A2	1108minus1GtoC	N/A	17	19568260	19568260
Sjoegren-Larsson syndrome	ALDH3A2	1108_1116delCTCATCAAAinsACAG | 1108_1116del9ins4 | 1108_1116delins	N/A	17	19568261	19568269
Sjoegren-Larsson syndrome	ALDH3A2	1139GtoA | S380N	rs72547574	17	19568292	19568292
Sjoegren-Larsson syndrome	ALDH3A2	1157AtoG | N386S	rs72547575	17	19568310	19568310
Febrile seizures	SEZ6	2206TtoA | Y736N	N/A	17	27285061	27285061
Febrile seizures	SEZ6	2182CtoA | L728I	N/A	17	27285085	27285085
Increased rigid-compulsive behaviour in autism	SLC6A4	1393TtoC | F465L	rs28914833	17	28537589	28537589
Autism, association with	SLC6A4	1273AtoC | I425L	rs28914832	17	28538374	28538374
Obsessive-compulsive disorder, susceptibility,	SLC6A4	1273AtoG | I425V	N/A	17	28538374	28538374
Neurofibromatosis 1	NF1	7285CtoT | R2429X	N/A	17	29677227	29677227
Neurofibromatosis 1	NF1	7285delC | 7285del1 | 7285del	N/A	17	29677227	29677227
Neurofibromatosis 1	NF1	7553minus2AtoG	N/A	17	29683476	29683476
Ovarian cancer, susceptibility to	RAD51D	345GtoC	N/A	17	33434385	33434385
Ovarian cancer, susceptibility to	RAD51D	270_271dupTA | 270_271dup2 | 270_271dup	N/A	17	33434458	33434459
Pancreas hypoplasia & multicystic renal dysplas	HNF1B	1406_1413dupTGCAGCCC | 1406_1413dup8 | 1406_1413dup	N/A	17	36061108	36061109
Cardiomyopathy, hypertrophic, association wi	TCAP	37_39delGAG | 37_39del3 | 37_39del	N/A	17	37821649	37821651
Cardiomyopathy, dilated	TCAP	53GtoA | R18Q	rs45614536	17	37821665	37821665
Muscular dystrophy, limb girdle 2G	TCAP	75GtoA | W25X	N/A	17	37821687	37821687
Cardiomyopathy, hypertrophic, association wi	TCAP	208CtoT | R70W	N/A	17	37822066	37822066
Osteogenesis imperfecta / Bruck syndrome	FKBP10	743dupC | 743dup1 | 743dup	N/A	17	39975477	39975478
Hyper-IgE syndrome	STAT3	2144plus1GtoA	N/A	17	40469199	40469199
STAT3 deficiency	STAT3	2141CtoT | T714I	N/A	17	40469203	40469203
Hyper-IgE syndrome	STAT3	2140AtoG | T714A	N/A	17	40469204	40469204
STAT3 deficiency	STAT3	2137GtoA | V713M	N/A	17	40469207	40469207
Hyper-IgE syndrome	STAT3	2137GtoC | V713L	N/A	17	40469207	40469207
Hyper-IgE syndrome	STAT3	2132TtoC | I711T	N/A	17	40469212	40469212
Hyper-IgE syndrome	STAT3	2129TtoG | F710C	N/A	17	40469215	40469215
Hyper-IgE syndrome	STAT3	2125AtoG | K709E	N/A	17	40469219	40469219
STAT3 deficiency	STAT3	2123CtoA | T708N	N/A	17	40469221	40469221
Hyper-IgE syndrome	STAT3	2123CtoG | T708S	N/A	17	40469221	40469221

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Hyper-IgE syndrome	STAT3	2116CtoA | L706M	N/A	17	40469228	40469228
Hyper-IgE syndrome	STAT3	2114AtoG | Y705C	N/A	17	40469230	40469230
Hyper-IgE syndrome	STAT3	1020_1022delGAC | 1020_1022del3 | 1020_1022del	N/A	17	40485718	40485720
Sanfilippo syndrome B	NAGLU	103CtoT | L35F	N/A	17	40688393	40688393
Sanfilippo syndrome B	NAGLU	112CtoT | R38W	N/A	17	40688402	40688402
Sanfilippo syndrome B	NAGLU	143TtoG | F48C	N/A	17	40688433	40688433
Sanfilippo syndrome B	NAGLU	144CtoA | F48L	rs104894599	17	40688434	40688434
Sanfilippo syndrome B	NAGLU	1674CtoG | Y558X	N/A	17	40695698	40695698
Sanfilippo syndrome B	NAGLU	1679TtoC | L560P	N/A	17	40695703	40695703
Sanfilippo syndrome B	NAGLU	1682TtoG | L561R	N/A	17	40695706	40695706
Breast cancer	BRCA1	4612CtoT | Q1538X	rs80356992	17	41226411	41226411
Breast cancer	BRCA1	4611_4612insG | 4611_4612ins1 | 4611_4612ins	rs80357915	17	41226411	41226412
Breast cancer	BRCA1	4609CtoT | Q1537X	rs80357229	17	41226414	41226414
Breast and/or ovarian cancer	BRCA1	4603GtoT | E1535X	rs80357366	17	41226420	41226420
Breast and/or ovarian cancer	BRCA1	4600GtoA | V1534M	rs55815649	17	41226423	41226423
Breast cancer	BRCA1	4595_4596insCT | 4595_4596ins2 | 4595_4596ins	rs80357699	17	41226427	41226428
Breast and/or ovarian cancer	BRCA1	4575_4585delAGAGGAGCTCA | 4575_4585del11 | 4575_4585del	N/A	17	41226438	41226448
Breast cancer	BRCA1	4574_4575delAA | 4574_4575del2 | 4574_4575del	rs80357813	17	41226448	41226449
Breast cancer	BRCA1	4565AtoG | Y1522C	rs80357379	17	41226458	41226458
Reduced promoter activity	PYY	NULL	rs2070592	17	42031331	42031331
Spherocytosis	SLC4A1	2608CtoT | R870W	rs28931585	17	42328574	42328574
Acanthocytosis	SLC4A1	2603CtoT | P868L	rs121912759	17	42328579	42328579
Distal renal tubular acidosis	SLC4A1	2573CtoA | A858D	rs121912751	17	42328609	42328609
Blood group variation	SLC4A1	2572GtoT | A858S	N/A	17	42328610	42328610
Distal renal tubular acidosis	SLC4A1	2548_2550delGTG | 2548_2550del3 | 2548_2550del	rs121912752	17	42328632	42328634
Spherocytosis	SLC4A1	2510CtoG | T837R	N/A	17	42328672	42328672
Spherocytosis	SLC4A1	2510CtoT | T837M	N/A	17	42328672	42328672
Spherocytosis	SLC4A1	2509AtoG | T837A	rs121912750	17	42328673	42328673
Spherocytosis	SLC4A1	2501AtoC | H834P	N/A	17	42328681	42328681
Amyotrophic lateral sclerosis	GRN	545CtoT | T182M	rs63750479	17	42427892	42427892
Glanzmann thrombasthenia	ITGA2B	2264GtoC | R755P	N/A	17	42454380	42454380
Glanzmann thrombasthenia	ITGA2B	2255TtoG | L752R	N/A	17	42454389	42454389
Glanzmann thrombasthenia	ITGA2B	2254CtoG | L752V	N/A	17	42454390	42454390
Glanzmann thrombasthenia	ITGA2B	2236GtoT | E746X	N/A	17	42454408	42454408
Glanzmann thrombasthenia	ITGA2B	2230dupG | 2230dup1 | 2230dup	N/A	17	42454413	42454414
Glanzmann thrombasthenia	ITGA2B	2153dupG | 2153dup1 | 2153dup	N/A	17	42455099	42455100
Glanzmann thrombasthenia	ITGA2B	959TtoC | F320S	N/A	17	42461301	42461301
Frontotemporal dementia	MAPT	50CtoT | T17M	rs144611688	17	44039753	44039753
Frontotemporal dementia	MAPT	224TtoC | V75A	N/A	17	44051754	44051754
Frontotemporal dementia	MAPT	256GtoA | G86S	rs63751135	17	44051786	44051786
Parkinson disease	MAPT	272CtoT | A91V	N/A	17	44051802	44051802
Tetra-amelia	WNT3	247CtoT | Q83X	rs104894653	17	44851109	44851109
Glanzmann thrombasthenia	ITGB3	812dupT | 812dup1 | 812dup	N/A	17	45364470	45364471
Glanzmann thrombasthenia	ITGB3	836AtoT | K279M	rs79775494	17	45364494	45364494
Glanzmann thrombasthenia 2	ITGB3	847_848delGC | 847_848del2 | 847_848del	N/A	17	45364505	45364506
Glanzmann thrombasthenia	ITGB3	856GtoA | G286R	N/A	17	45364514	45364514
Glanzmann thrombasthenia 2	ITGB3	863TtoC | L288P	N/A	17	45364521	45364521
Glanzmann thrombasthenia	ITGB3	2302minus3CtoG	N/A	17	45387502	45387502
Glanzmann thrombasthenia	ITGB3	2315TtoC | L772P	N/A	17	45387518	45387518
Glanzmann thrombasthenia	ITGB3	2332TtoC | S778P	rs121918447	17	45387535	45387535
Diabetes, type 1, association with	TBX21	99CtoG | H33Q	rs2240017	17	45810919	45810919
Osteogenesis imperfecta I	COL1A1	2613delT | 2613del1 | 2613del	rs72653144	17	48267220	48267220
Osteogenesis imperfecta II	COL1A1	2605GtoT | G869C	rs72653143	17	48267228	48267228
Connective tissue weakness	COL1A1	2387GtoA | R796H	rs72651662	17	48267914	48267914
Osteogenesis imperfecta	COL1A1	1984minus5CtoA	rs66592376	17	48269390	48269390
Osteogenesis imperfecta I	COL1A1	1984-5delC | 1984-5del1 | 1984-5del	N/A	17	48269390	48269390
Osteogenesis imperfecta	COL1A1	1982_1983delAGinsTT | 1982_1983del2ins2 | 1982_1983delins	N/A	17	48269836	48269837
Osteogenesis imperfecta	COL1A1	1976GtoC | G659A	rs72651630	17	48269843	48269843
Osteogenesis imperfecta	COL1A1	1310GtoA | G437D	N/A	17	48272451	48272451
Osteogenesis imperfecta II	COL1A1	1301GtoT | G434V	rs72648333	17	48272460	48272460
Osteogenesis imperfecta I	COL1A1	543plus2TtoC	N/A	17	48275792	48275792
Ehlers-Danlos syndrome VII	COL1A1	543GtoA	N/A	17	48275794	48275794
Osteogenesis imperfecta I	COL1A1	533delC | 533del1 | 533del	N/A	17	48275804	48275804
Osteogenesis imperfecta I	COL1A1	484delC | 484del1 | 484del	N/A	17	48275853	48275853
Ehlers-Danlos syndrome VII	COL1A1	472minus1GtoA	rs72667020	17	48275866	48275866
Osteogenesis imperfecta I	COL1A1	472minus2AtoG	rs72667019	17	48275867	48275867
Mulibrey nanism	TRIM37	1894_1895delGA | 1894_1895del2 | 1894_1895del	N/A	17	57109310	57109311
Short stature	GH1	426CtoG | D142E	N/A	17	61995150	61995150
Short stature	GH1	413AtoG | D138G	rs137853221	17	61995163	61995163
Lung cancer, susceptibility to, association with	GH1	406GtoA | V136I	rs5388	17	61995170	61995170
Growth hormone deficiency	GH1	406GtoT | V136F	N/A	17	61995170	61995170
Growth hormone deficiency	GH1	400AtoC | S134R	N/A	17	61995176	61995176
Growth hormone deficiency	GH1	400AtoT | S134C	N/A	17	61995176	61995176
Growth hormone deficiency	GH1	352TtoC | F118L	N/A	17	61995224	61995224
Short stature	GH1	350AtoT | Q117L	N/A	17	61995226	61995226
Growth hormone deficiency	GH1	344CtoT | P115L	N/A	17	61995232	61995232
Paramyotonia congenita, von Eulenburg	SCN4A	4367GtoA | G1456E	rs121908554	17	62019275	62019275
Periodic paralysis	SCN4A	4351CtoT | R1451C	N/A	17	62019291	62019291
Paramyotonia congenita	SCN4A	4343GtoA | R1448H	rs121908545	17	62019299	62019299
Myotonia	SCN4A	4343GtoC | R1448P	N/A	17	62019299	62019299
Paramyotonia congenita	SCN4A	4343GtoT | R1448L	N/A	17	62019299	62019299
Paramyotonia congenita	SCN4A	4342CtoA | R1448S	N/A	17	62019300	62019300
Paramyotonia congenita	SCN4A	4342CtoT | R1448C	rs121908544	17	62019300	62019300
Carney complex	PRKAR1A	178minus2AtoG	N/A	17	66518895	66518895
Carney complex	PRKAR1A	178minus1GtoA	N/A	17	66518896	66518896
Carney complex	PRKAR1A	187AtoT | K63X	N/A	17	66518906	66518906
Carney complex	PRKAR1A	190CtoT | Q64X	N/A	17	66518909	66518909
Carney complex	PRKAR1A	205CtoT | Q69X	N/A	17	66518924	66518924
Carney complex	PRKAR1A	206AtoG | Q69R	N/A	17	66518925	66518925
Carney complex	PRKAR1A	220CtoT | R74C	rs137853303	17	66518939	66518939
Carney complex	PRKAR1A	220_221delCG | 220_221del2 | 220_221del	N/A	17	66518939	66518940
Cushing syndrome	PRKAR1A	221_230delGTACAGACTC | 221_230del10 | 221_230del	N/A	17	66518940	66518949
Carney complex	PRKAR1A	242delA | 242del1 | 242del	N/A	17	66518961	66518961
Carney complex	PRKAR1A	267delA | 267del1 | 267del	N/A	17	66518986	66518986
Carney complex	PRKAR1A	279_282delTAGG | 279_282del4 | 279_282del	N/A	17	66518998	66519001

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Carney complex	PRKAR1A	286CtoT | R96X	N/A	17	66519005	66519005
Amelogenesis imperfecta & gingival hyperplas	FAM20A	1175_1179delGGCTC | 1175_1179del5 | 1175_1179del	N/A	17	66537030	66537034
Amelogenesis imperfecta & gingival hyperplas	FAM20A	34_35delCT | 34_35del2 | 34_35del	N/A	17	66596773	66596774
Andersen-Tawil syndrome	KCNJ2	118CtoT | R40X	N/A	17	68171298	68171298
Campomelic dysplasia	SOX9	1061_1069delCGGCCCCGC | 1061_1069del9 | 1061_1069del	N/A	17	70120059	70120067
Ciliary dyskinesia, primary	DNAI2	346minus3TtoG	N/A	17	72283113	72283113
Usher syndrome, atypical	USH1G	163_164+13delGGGTGAGTACGCCCC | 163_164+13del15 | 163_164+13del	N/A	17	72918992	72919006
Usher syndrome 1	USH1G	84dupC | 84dup1 | 84dup	N/A	17	72919084	72919085
Usher syndrome 1	USH1G	46CtoG | L16V	N/A	17	72919123	72919123
Pontocerebellar hypoplasia 4	TSEN54	1138GtoT | E380X	N/A	17	73518300	73518300
Pontocerebellar hypoplasia 4	TSEN54	1172_1185delAGAGGAGCCAGCGC | 1172_1185del14 | 1172_1185del	N/A	17	73518334	73518347
Epidermolysis bullosa with pyloric atresia	ITGB4	391GtoT | D131Y	N/A	17	73723858	73723858
Epidermolysis bullosa with pyloric atresia	ITGB4	818GtoA | G273D	N/A	17	73726401	73726401
Haemophagocytic lymphohistiocytosis, familia	UNC13D	3037dupG | 3037dup1 | 3037dup	N/A	17	73824981	73824982
Haemophagocytic lymphohistiocytosis, familia	UNC13D	2783GtoC | R928P	N/A	17	73826490	73826490
Haemophagocytic lymphohistiocytosis, familia	UNC13D	2782CtoT | R928C	rs35037984	17	73826491	73826491
Haemophagocytic lymphohistiocytosis, familia	UNC13D	2709plus1GtoA	N/A	17	73826658	73826658
Haemophagocytic lymphohistiocytosis, familia	UNC13D	2437_2439delAACinsT | 2437_2439del3ins1 | 2437_2439delins	N/A	17	73829055	73829057
Haemophagocytic lymphohistiocytosis	UNC13D	2381delT | 2381del1 | 2381del	N/A	17	73829113	73829113
Haemophagocytic lymphohistiocytosis, familia	UNC13D	1847AtoG | E616G	N/A	17	73831491	73831491
Haemophagocytic lymphohistiocytosis, familia	UNC13D	1847AtoG	N/A	17	73831491	73831491
Haemophagocytic lymphohistiocytosis, familia	UNC13D	1828_1839delCGCGCTGTGCAG | 1828_1839del12 | 1828_1839del	N/A	17	73831499	73831510
Haemophagocytic lymphohistiocytosis, familia	UNC13D	1828_1829insA | 1828_1829ins1 | 1828_1829ins	N/A	17	73831509	73831510
Peroxisomal acyl-CoA oxidase deficiency	ACOX1	1872TtoA | Y624X	N/A	17	73944395	73944395
Peroxisomal acyl-CoA oxidase deficiency	ACOX1	1789_1792delACTC | 1789_1792del4 | 1789_1792del	N/A	17	73944475	73944478
Epidermodysplasia verruciformis	TMC6	968delT | 968del1 | 968del	N/A	17	76120184	76120184
Epidermodysplasia verruciformis	TMC6	744CtoA | Y248X	rs121908329	17	76120752	76120752
Glycogen storage disease 2	GAA	2841dupT | 2841dup1 | 2841dup	N/A	17	78093112	78093113
Psoriasis	CARD14	571GtoT | V191L	N/A	17	78157933	78157933
Psoriasis, association with	CARD14	599GtoA | S200N	N/A	17	78157961	78157961
Psoriasis	CARD14	854AtoG | D285G	N/A	17	78163562	78163562
Sanfilippo syndrome A	SGSH	1465GtoT | E489X	N/A	17	78184295	78184295
Sanfilippo syndrome A	SGSH	1456GtoT | V486F	N/A	17	78184304	78184304
Sanfilippo syndrome A	SGSH	739GtoC | D247H	N/A	17	78187609	78187609
Sanfilippo syndrome A	SGSH	739GtoT | D247Y	N/A	17	78187609	78187609
Sanfilippo syndrome A	SGSH	734GtoA | R245H	rs104894635	17	78187614	78187614
Sanfilippo syndrome A	SGSH	35TtoA | L12Q	N/A	17	78194078	78194078
Sanfilippo syndrome A	SGSH	35TtoC | L12P	N/A	17	78194078	78194078
Sanfilippo syndrome A	SGSH	29dupC | 29dup1 | 29dup	N/A	17	78194083	78194084
Sanfilippo syndrome A	SGSH	2TtoC | M1T	N/A	17	78194111	78194111
Sanfilippo syndrome A	SGSH	1AtoC | M1L	N/A	17	78194112	78194112
Deafness, dominant progressive	ACTG1	994CtoG | P332A	N/A	17	79477850	79477850
Adenomatous polyposis coli	CSNK1D	971GtoA | R324H	N/A	17	80207393	80207393
Transcriptional activity, association with	TYMS	NULL	rs2853542	18	657685	657685
Schizophrenia, association with	IMPA2	558CtoT | F186F	rs2075825	18	12028109	12028109
Glucocorticoid deficiency	MC2R	866dupA | 866dup1 | 866dup	N/A	18	13884651	13884652
Niemann-Pick disease C	NPC1	2245plus1GtoA	N/A	18	21123418	21123418
Niemann-Pick disease C	NPC1	2240delT | 2240del1 | 2240del	N/A	18	21123424	21123424
Niemann-Pick disease C	NPC1	2234CtoA | A745E	N/A	18	21123430	21123430
Niemann-Pick disease C	NPC1	2224GtoA | E742K	N/A	18	21123440	21123440
Niemann-Pick disease C	NPC1	2217_2222delCTTTTC | 2217_2222del6 | 2217_2222del	N/A	18	21123442	21123447
Niemann-Pick disease C	NPC1	2213CtoA | S738X	N/A	18	21123451	21123451
Niemann-Pick disease C	NPC1	1529AtoC | H510P	N/A	18	21134746	21134746
Niemann-Pick disease C	NPC1	1526AtoC | Y509S	N/A	18	21134749	21134749
Niemann-Pick disease C	NPC1	1526AtoG | Y509C	N/A	18	21134749	21134749
Niemann-Pick disease C	NPC1	852delT | 852del1 | 852del	N/A	18	21140224	21140224
Niemann-Pick disease C	NPC1	275AtoG | Q92R	N/A	18	21152050	21152050
Niemann-Pick disease C	NPC1	221GtoA | C74Y	N/A	18	21152104	21152104
Niemann-Pick disease C	NPC1	47_48delGT | 47_48del2 | 47_48del	N/A	18	21166260	21166261
Niemann-Pick disease C	NPC1	7GtoA | A3T	N/A	18	21166301	21166301
Epidermolysis bullosa, junctional	LAMA3	4335dupA | 4335dup1 | 4335dup	N/A	18	21523887	21523888
Epidermolysis bullosa, Herlitz	LAMA3	4383plus1GtoA	N/A	18	21523936	21523936
Striate palmoplantar keratoderma	DSG1	1931delA | 1931del1 | 1931del	N/A	18	28925992	28925992
Hypotrichosis	DSG4	574TtoC | S192P	N/A	18	28970675	28970675
Autism	KATNAL2	510plus1GtoA	N/A	18	44589733	44589733
Microvillus inclusion disease	MYO5B	2330delG | 2330del1 | 2330del	N/A	18	47432873	47432873
Microvillus inclusion disease	MYO5B	2004minus2AtoG	N/A	18	47455970	47455970
Microvillus inclusion disease	MYO5B	1AtoG | M1V	N/A	18	47721163	47721163
Intrahepatic cholestasis, familial progressive	ATP8B1	2941GtoA | E981K	N/A	18	55321298	55321298
Anophthalmia	RAX	664delT | 664del1 | 664del	N/A	18	56936613	56936613
Anophthalmia and sclerocornea	RAX	575GtoA | R192Q	rs121909127	18	56936702	56936702
Expansile skeletal hyperphosphatasia	TNFRSF11A	48_62dupGCTCTGCGCGCTGCT | 48_62dup15 | 48_62dup	N/A	18	59992647	59992648
Familial expansile osteolysis	TNFRSF11A	45_62dupGCTGCTCTGCGCGCTGCT | 45_62dup18 | 45_62dup	N/A	18	59992647	59992648
Familial expansile osteolysis	TNFRSF11A	46_63dupCTGCTCTGCGCGCTGCTC | 46_63dup18 | 46_63dup	N/A	18	59992648	59992649
Osteopetrosis, RANK-dependent, autosomal re	TNFRSF11A	372CtoA | C124X	N/A	18	60021712	60021712
Altered splicing	HCN2	1584plus7CtoT	rs55978915	19	610412	610412
Peutz-Jeghers syndrome	STK11	47AtoG | E16G	N/A	19	1206959	1206959
Peutz-Jeghers syndrome	STK11	77_78insACAT | 77_78ins4 | 77_78ins	N/A	19	1206989	1206990
Peutz-Jeghers syndrome	STK11	109CtoT | Q37X	rs121913324	19	1207021	1207021
Peutz-Jeghers syndrome	STK11	111delG | 111del1 | 111del	N/A	19	1207023	1207023
Peutz-Jeghers syndrome	STK11	115_125delCGCCGCAAGCG | 115_125del11 | 115_125del	N/A	19	1207027	1207037
Peutz-Jeghers syndrome	STK11	291minus2AtoC	N/A	19	1218414	1218414
Peutz-Jeghers syndrome	STK11	291minus2AtoG	N/A	19	1218414	1218414
Peutz-Jeghers syndrome	STK11	291minus2AtoT	N/A	19	1218414	1218414
Peutz-Jeghers syndrome	STK11	291minus1GtoC	rs112675807	19	1218415	1218415
Peutz-Jeghers syndrome	STK11	291minus1GtoT	N/A	19	1218415	1218415
Peutz-Jeghers syndrome	STK11	351dupA | 351dup1 | 351dup	N/A	19	1218476	1218477
Peutz-Jeghers syndrome	STK11	353dupA | 353dup1 | 353dup	N/A	19	1218478	1218479
Peutz-Jeghers syndrome	STK11	354CtoA | Y118X	N/A	19	1218479	1218479
Peutz-Jeghers syndrome	STK11	354CtoG | Y118X	N/A	19	1218479	1218479
Peutz-Jeghers syndrome	STK11	367CtoT | Q123X	N/A	19	1218492	1218492
Peutz-Jeghers syndrome	STK11	369delG | 369del1 | 369del	N/A	19	1218494	1218494
Peutz-Jeghers syndrome	STK11	374plus1AtoG	N/A	19	1218500	1218500
Peutz-Jeghers syndrome	STK11	663_664insGG | 663_664ins2 | 663_664ins	N/A	19	1220645	1220646
Peutz-Jeghers syndrome	STK11	666delC | 666del1 | 666del	N/A	19	1220648	1220648

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Peutz-Jeghers syndrome	STK11	667_668insGG | 667_668ins2 | 667_668ins	N/A	19	1220649	1220650
Peutz-Jeghers syndrome	STK11	688AtoC | T230P	N/A	19	1220670	1220670
Peutz-Jeghers syndrome	STK11	921-7_921delTTTGCAGCinsCAG | 921-7_921del8ins3 | 921-7_921delins	N/A	19	1222977	1222984
Peutz-Jeghers syndrome	STK11	921minus2AtoG	N/A	19	1222982	1222982
Peutz-Jeghers syndrome	STK11	920plus978GtoC	N/A	19	1222983	1222983
Peutz-Jeghers syndrome	STK11	921minus1GtoC	N/A	19	1222983	1222983
Peutz-Jeghers syndrome	STK11	923GtoA | W308X	N/A	19	1222986	1222986
Peutz-Jeghers syndrome	STK11	924GtoT | W308C	N/A	19	1222987	1222987
Cardiomyopathy, neonatal	NDUFS7	1-3_1delAAGAinsTCT | 1-3_1del4ins3 | 1-3_1delins	N/A	19	1383923	1383926
Guanidinoacetate methyltransferase deficienc	GAMT	160GtoC | A54P	N/A	19	1401316	1401316
Guanidinoacetate methyltransferase deficienc	GAMT	152AtoC | H51P	N/A	19	1401324	1401324
Guanidinoacetate methyltransferase deficienc	GAMT	148AtoC | M50L	rs104894694	19	1401328	1401328
Guanidinoacetate methyltransferase deficienc	GAMT	64dupG | 64dup1 | 64dup	N/A	19	1401411	1401412
Guanidinoacetate methyltransferase deficienc	GAMT	59GtoC | W20S	rs80338734	19	1401417	1401417
Autism	TCF3	1382dupA | 1382dup1 | 1382dup	N/A	19	1619177	1619178
Persistent Mullerian ducts syndrome	AMH	974AtoG | Q325R	rs140765565	19	2251247	2251247
Persistent Mullerian ducts syndrome	AMH	977GtoT | G326V	N/A	19	2251250	2251250
Persistent Mullerian ducts syndrome	AMH	1144GtoT | E382X	N/A	19	2251417	2251417
Persistent Mullerian ducts syndrome	AMH	1213GtoC | A405P	N/A	19	2251486	2251486
Reduced blood pressure, association with	TBXA2R	NULL	rs13306046	19	3595522	3595522
Haemolytic uraemic syndrome, atypical	C3	3187AtoC | S1063R	N/A	19	6693466	6693466
Complement C3 deficiency	C3	1648TtoC | S550P	N/A	19	6710688	6710688
Complement C3 deficiency	C3	1645GtoA | D549N	N/A	19	6710691	6710691
Insulin resistance	INSR	1372AtoG | N458D	rs121913160	19	7170659	7170659
Leprechaunism	INSR	1316GtoC | W439S	rs121913158	19	7170715	7170715
Insulin resistance	INSR	22_31delGGGGCGGCGG | 22_31del10 | 22_31del	N/A	19	7293872	7293881
Hemophagocytic lymphohistiocytosis type 5	STXBP2	474_483delCCCCTTCCGGinsGA | 474_483del10ins2 | 474_483delins	N/A	19	7706635	7706644
Lower plasma triglyceride level	ANGPTL4	80CtoG | P27R	N/A	19	8429285	8429285
Lower plasma triglyceride level	ANGPTL4	229GtoA | G77R	N/A	19	8429434	8429434
Sensorineural hearing loss, bilateral	MYO1F	1656GtoC | K552N	N/A	19	8604867	8604867
Weill-Marchesani syndrome	ADAMTS10	952CtoT | Q318X	rs121434359	19	8661959	8661959
Coffin-Siris syndrome	SMARCA4	3469CtoG | R1157G	N/A	19	11141492	11141492
Hypercholesterolaemia	LDLR	1587minus2AtoG	N/A	19	11226768	11226768
Hypercholesterolaemia	LDLR	1587minus2AtoT	N/A	19	11226768	11226768
Hypercholesterolaemia	LDLR	1587minus1GtoA	N/A	19	11226769	11226769
Hypercholesterolaemia	LDLR	1587_1591delCTTCA | 1587_1591del5 | 1587_1591del	N/A	19	11226770	11226774
Hypercholesterolaemia	LDLR	1592TtoA | M531K	N/A	19	11226775	11226775
Hypercholesterolaemia	LDLR	1597TtoC | W533R	N/A	19	11226780	11226780
Hypercholesterolaemia	LDLR	1598GtoA | W533X	N/A	19	11226781	11226781
Hypercholesterolaemia	LDLR	1599GtoA | W533X	N/A	19	11226782	11226782
Hypercholesterolaemia	LDLR	1600_1608delACTGACTGG | 1600_1608del9 | 1600_1608del	N/A	19	11226783	11226791
Hypercholesterolaemia	LDLR	1601CtoA | T534N	N/A	19	11226784	11226784
Hypercholesterolaemia	LDLR	1603GtoT | D535Y	N/A	19	11226786	11226786
Hypercholesterolaemia	LDLR	1606TtoG | W536G	N/A	19	11226789	11226789
Hypercholesterolaemia	LDLR	1607GtoA | W536X	N/A	19	11226790	11226790
Hypercholesterolaemia	LDLR	1609GtoT | G537X	N/A	19	11226792	11226792
Hypercholesterolaemia	LDLR	1610delG | 1610del1 | 1610del	N/A	19	11226793	11226793
Hypercholesterolaemia	LDLR	1612AtoT | T538S	N/A	19	11226795	11226795
Hypercholesterolaemia	LDLR	1618GtoA | A540T	N/A	19	11226801	11226801
Hypercholesterolaemia	LDLR	1618GtoT | A540S	N/A	19	11226801	11226801
Hypercholesterolaemia	LDLR	1618delG | 1618del1 | 1618del	N/A	19	11226801	11226801
Hypercholesterolaemia	LDLR	1625TtoG | I542S	N/A	19	11226808	11226808
Hypercholesterolaemia	LDLR	1632delA | 1632del1 | 1632del	N/A	19	11226815	11226815
Hypercholesterolaemia	LDLR	1633GtoA | G545R	N/A	19	11226816	11226816
Hypercholesterolaemia	LDLR	1633GtoT | G545W	N/A	19	11226816	11226816
Hypercholesterolaemia	LDLR	1636GtoC | G546R	N/A	19	11226819	11226819
Hypercholesterolaemia	LDLR	1637GtoA | G546D	rs28942081	19	11226820	11226820
Hypercholesterolaemia	LDLR	1637GtoT | G546V	N/A	19	11226820	11226820
Hypercholesterolaemia	LDLR	1640TtoC | L547P	N/A	19	11226823	11226823
Hypercholesterolaemia	LDLR	1640_1652delTGAATGGTGTGGAinsAGCGTCATCTTCCTGAC | 1640_1652del13ins17 | 1640_1652delins	N/A	19	11226823	11226835
Hypercholesterolaemia	LDLR	1644TtoG | N548K	N/A	19	11226827	11226827
Hypercholesterolaemia	LDLR	1644_1645insC | 1644_1645ins1 | 1644_1645ins	N/A	19	11226827	11226828
Hypercholesterolaemia	LDLR	1646GtoA | G549D	rs28941776	19	11226829	11226829
Hypercholesterolaemia	LDLR	1646GtoT | G549V	N/A	19	11226829	11226829
Hypercholesterolaemia	LDLR	1651delG | 1651del1 | 1651del	N/A	19	11226834	11226834
Hypercholesterolaemia	LDLR	1655delT | 1655del1 | 1655del	N/A	19	11226838	11226838
Hypercholesterolaemia	LDLR	1658AtoG | Y553C	N/A	19	11226841	11226841
Hypercholesterolaemia	LDLR	1659_1661delCTCinsATACTTTCA | 1659_1661del3ins9 | 1659_1661delins	N/A	19	11226842	11226844
Hypercholesterolaemia	LDLR	1661CtoA | S554X	N/A	19	11226844	11226844
Hypercholesterolaemia	LDLR	1661CtoT | S554L	N/A	19	11226844	11226844
Hypercholesterolaemia	LDLR	1687CtoT | P563S	N/A	19	11226870	11226870
Hypercholesterolaemia	LDLR	1688CtoA | P563H	N/A	19	11226871	11226871
Hypercholesterolaemia	LDLR	1689dupC | 1689dup1 | 1689dup	N/A	19	11226872	11226873
Hypercholesterolaemia	LDLR	1690AtoC | N564H	N/A	19	11226873	11226873
Hypercholesterolaemia	LDLR	1690AtoG | N564D	N/A	19	11226873	11226873
Hypercholesterolaemia	LDLR	1691AtoG | N564S	N/A	19	11226874	11226874
Hypercholesterolaemia	LDLR	1694GtoC | G565A	N/A	19	11226877	11226877
Hypercholesterolaemia	LDLR	1694GtoT | G565V	N/A	19	11226877	11226877
Hypercholesterolaemia	LDLR	1698_1704delCACCCTAinsGCCCAAT | 1698_1704del7ins7 | 1698_1704delins	N/A	19	11226881	11226887
Hypercholesterolaemia	LDLR	1702CtoG | L568V	N/A	19	11226885	11226885
Hypercholesterolaemia	LDLR	1702delC | 1702del1 | 1702del	N/A	19	11226885	11226885
Hypercholesterolaemia	LDLR	1703TtoC | L568P	N/A	19	11226886	11226886
Hypercholesterolaemia	LDLR	1705GtoT | D569Y	N/A	19	11226888	11226888
Hypercholesterolaemia	LDLR	1702_1705dupCTAG | 1702_1705dup4 | 1702_1705dup	N/A	19	11226888	11226889
Hypercholesterolaemia	LDLR	1705plus1GtoA	N/A	19	11226889	11226889
Hypercholesterolaemia	LDLR	1705plus1GtoC	N/A	19	11226889	11226889
Hypercholesterolaemia	LDLR	1705plus1GtoT	N/A	19	11226889	11226889
Hypercholesterolaemia	LDLR	1705+1delG | 1705+1del1 | 1705+1del	N/A	19	11226889	11226889
Hypercholesterolaemia	LDLR	1705+2_1705+3insC | 1705+2_1705+3ins1 | 1705+2_1705+3ins	N/A	19	11226890	11226891
Polycystic liver disease	PRKCSH	464AtoG | K155R	N/A	19	11552168	11552168
Polycystic liver disease	PRKCSH	466CtoT | Q156X	N/A	19	11552170	11552170
Polycystic liver disease	PRKCSH	487CtoT | Q163X	N/A	19	11553219	11553219
Polycystic liver disease	PRKCSH	668delA | 668del1 | 668del	N/A	19	11556273	11556273
Mannosidosis, alpha	MAN2B1	1390CtoT | Q464X	N/A	19	12768289	12768289
Mannosidosis, alpha	MAN2B1	1388_1389delGC | 1388_1389del2 | 1388_1389del	N/A	19	12768290	12768291

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Mannosidosis, alpha	MAN2B1	1383CtoA | Y461X	N/A	19	12768296	12768296
Mannosidosis, alpha	MAN2B1	1383CtoG | Y461X	N/A	19	12768296	12768296
Mannosidosis, alpha	MAN2B1	1370TtoG | V457G	N/A	19	12768309	12768309
Mannosidosis, alpha	MAN2B1	1358CtoA | S453Y	N/A	19	12768321	12768321
Mannosidosis, alpha	MAN2B1	1358CtoT | S453F	N/A	19	12768321	12768321
Mannosidosis, alpha	MAN2B1	1351GtoT | G451C	N/A	19	12768328	12768328
Mannosidosis, alpha	MAN2B1	1340AtoT | D447V	N/A	19	12768339	12768339
Mannosidosis, alpha	MAN2B1	1333CtoT | H445Y	N/A	19	12768346	12768346
Mannosidosis, alpha	MAN2B1	157GtoT | E53X	N/A	19	12777359	12777359
Mannosidosis, alpha	MAN2B1	152delG | 152del1 | 152del	N/A	19	12777364	12777364
Increased Hb F levels	KLF1	526_527insCGGCGCC | 526_527ins7 | 526_527ins	N/A	19	12996517	12996518
Glutaricacidaemia I	GCDH	148TtoC | W50R	N/A	19	13002665	13002665
Glutaricacidaemia I	GCDH	150GtoC | W50C	N/A	19	13002667	13002667
Glutaricacidaemia I	GCDH	157CtoT | P53S	N/A	19	13002674	13002674
Glutaricacidaemia I	GCDH	158delC | 158del1 | 158del	N/A	19	13002675	13002675
Glutaricacidaemia I	GCDH	877GtoA | A293T	rs121434371	19	13007748	13007748
Glutaricacidaemia I	GCDH	877delG | 877del1 | 877del	N/A	19	13007748	13007748
Glutaricacidaemia I	GCDH	880CtoT | R294W	N/A	19	13007751	13007751
Glutaricacidaemia I	GCDH	883TtoC | Y295H	rs121434366	19	13007754	13007754
Glutaricacidaemia I	GCDH	905TtoC | L302P	N/A	19	13007776	13007776
Glutaricacidaemia I	GCDH	910GtoA | A304T	N/A	19	13007781	13007781
Glutaricacidaemia I	GCDH	914CtoT | S305L	N/A	19	13007785	13007785
Glutaricacidaemia I	GCDH	923GtoC | C308S	N/A	19	13007794	13007794
Glutaricacidaemia I	GCDH	926TtoG | L309W	N/A	19	13007797	13007797
Glutaricacidaemia I	GCDH	937CtoT | R313W	N/A	19	13007808	13007808
Glutaricacidaemia I	GCDH	938GtoA | R313Q	N/A	19	13007809	13007809
Glutaricacidaemia I	GCDH	947CtoA | A316D	N/A	19	13007818	13007818
Episodic ataxia 2	CACNA1A	5338CtoT | R1780X	N/A	19	13342589	13342589
CADASIL	NOTCH3	3944GtoA | C1315Y	N/A	19	15288795	15288795
White matter lesions	NOTCH3	3785GtoT | R1262L	N/A	19	15289686	15289686
CADASIL	NOTCH3	1630CtoT | R544C	N/A	19	15298126	15298126
CADASIL	NOTCH3	1625GtoA | C542Y	N/A	19	15298131	15298131
CADASIL	NOTCH3	1582GtoT | G528C	N/A	19	15298716	15298716
CADASIL	NOTCH3	1532GtoA | C511Y	N/A	19	15298766	15298766
CADASIL	NOTCH3	1532GtoT | C511F	N/A	19	15298766	15298766
CADASIL	NOTCH3	1531TtoC | C511R	N/A	19	15298767	15298767
CADASIL	NOTCH3	194GtoA | C65Y	N/A	19	15308314	15308314
CADASIL	NOTCH3	194GtoC | C65S	N/A	19	15308314	15308314
CADASIL	NOTCH3	193TtoG | C65G	N/A	19	15308315	15308315
CADASIL	NOTCH3	181CtoT | R61W	N/A	19	15308327	15308327
CADASIL	NOTCH3	179CtoG | S60C	N/A	19	15308329	15308329
CADASIL	NOTCH3	160CtoT | R54C	N/A	19	15308348	15308348
CADASIL	NOTCH3	157GtoT | G53C	N/A	19	15308351	15308351
CADASIL	NOTCH3	146GtoA | C49Y	N/A	19	15308362	15308362
CADASIL	NOTCH3	146GtoT | C49F	N/A	19	15308362	15308362
CADASIL	NOTCH3	145TtoC | C49R	N/A	19	15308363	15308363
CADASIL	NOTCH3	145TtoG | C49G	N/A	19	15308363	15308363
CADASIL	NOTCH3	128GtoT | C43F	N/A	19	15308380	15308380
CADASIL	NOTCH3	127TtoG | C43G	N/A	19	15308381	15308381
Immunodeficiency, severe combined	JAK3	1767CtoT	N/A	19	17947957	17947957
Immunodeficiency, severe combined	JAK3	1765GtoA | G589S	N/A	19	17947959	17947959
IL12RB1 deficiency	IL12RB1	1561CtoG | R521G	N/A	19	18174743	18174743
IL12RB1 deficiency	IL12RB1	1561CtoT | R521X	N/A	19	18174743	18174743
IL12RB1 deficiency	IL12RB1	557GtoA | C186Y	N/A	19	18187130	18187130
IL12RB1 deficiency	IL12RB1	557GtoC | C186S	N/A	19	18187130	18187130
Mycobacterial infection	IL12RB1	550minus2AtoG	N/A	19	18187139	18187139
Mycobacterial disease, mendelian susceptibili	IL12RB1	301GtoT | D101Y	N/A	19	18191750	18191750
IL12RB1 deficiency	IL12RB1	199GtoT | E67X	N/A	19	18193000	18193000
Congenital heart defects	GDF1	934CtoA | P312T	N/A	19	18979591	18979591
Congenital heart defects	GDF1	925TtoC | S309P	N/A	19	18979600	18979600
Right atrial isomerism	GDF1	793_803delGGCGCTTGTCG | 793_803del11 | 793_803del	N/A	19	18979722	18979732
Congenital heart defects	GDF1	800GtoA | C267Y	rs121434423	19	18979725	18979725
Congenital heart defects	GDF1	784GtoA | G262S	N/A	19	18979741	18979741
Congenital heart defects	GDF1	681CtoA | C227X	rs121434422	19	18979844	18979844
Retinal dysfunction	RGS9BP	94_102delGACTCGCAG | 94_102del9 | 94_102del	N/A	19	33167263	33167271
Cystinuria	SLC7A9	279_280delAG | 279_280del2 | 279_280del	N/A	19	33355200	33355201
Cystinuria	SLC7A10	336GtoC | E112D	rs79717007	19	33706695	33706695
Prolidase deficiency	PEPD	692_694delACT | 692_694del3 | 692_694del	N/A	19	33904527	33904529
Glucosephosphate isomerase deficiency	GPI	59AtoC | H20P	rs137853586	19	34856230	34856230
Minimal change nephrotic syndrome	NPHS1	2635GtoA | G879R	N/A	19	36333054	36333054
Nephrotic syndrome	NPHS1	2633dupA | 2633dup1 | 2633dup	N/A	19	36333055	36333056
Congenital nephrotic syndrome, Finnish type	NPHS1	2614delG | 2614del1 | 2614del	N/A	19	36333075	36333075
Congenital nephrotic syndrome, Finnish type	NPHS1	2606_2607dupCC | 2606_2607dup2 | 2606_2607dup	N/A	19	36333081	36333082
Congenital nephrotic syndrome, Finnish type	NPHS1	2500GtoT | V834F	N/A	19	36333287	36333287
Nephrotic syndrome	NPHS1	2495TtoC | L832P	N/A	19	36333292	36333292
Diffuse mesangial sclerosis	NPHS1	2386GtoA | G796R	N/A	19	36333401	36333401
Minimal change nephrotic syndrome	NPHS1	2357GtoA | S786N	N/A	19	36333430	36333430
Congenital nephrotic syndrome, Finnish type	NPHS1	2156_2163delTGCACTGC | 2156_2163del8 | 2156_2163del	N/A	19	36335054	36335061
Congenital nephrotic syndrome, Finnish type	NPHS1	2160dupC | 2160dup1 | 2160dup	N/A	19	36335056	36335057
Nephrotic syndrome	NPHS1	2149TtoG | Y717D	N/A	19	36335068	36335068
Focal segmental glomerulosclerosis	NPHS1	2143GtoC | G715R	N/A	19	36335074	36335074
Nephrotic syndrome	NPHS1	2131CtoA | R711S	N/A	19	36335086	36335086
Congenital nephrotic syndrome, Finnish type	NPHS1	2131CtoT | R711C	N/A	19	36335086	36335086
Nephrotic syndrome	NPHS1	2126TtoG | V709G	N/A	19	36335091	36335091
Nephrotic syndrome	NPHS1	2104GtoT | G702W	N/A	19	36335113	36335113
Nephrotic syndrome	NPHS1	2072minus6CtoG	N/A	19	36335151	36335151
Nephrotic syndrome	NPHS1	58plus1GtoT	N/A	19	36342681	36342681
Microcephaly, primary autosomal recessive 2	WDR62	1313GtoA | R438H	N/A	19	36572414	36572414
Sodium diarrhoea	SPINT2	553plus2TtoA	N/A	19	38780922	38780922
Central core disease	RYR1	1422GtoT | Q474H	N/A	19	38943636	38943636
Myopathy, congenital with cores	RYR1	1739_1742dupATCA | 1739_1742dup4 | 1739_1742dup	N/A	19	38946342	38946343
Malignant hyperthermia	RYR1	3127CtoT | R1043C	rs111272095	19	38956987	38956987
Malignant hyperthermia	RYR1	4178AtoG | K1393R	rs137933390	19	38965975	38965975
Myopathy, congenital	RYR1	4816CtoT | R1606C	N/A	19	38974038	38974038
Myopathy, congenital	RYR1	5726_5727delAG | 5726_5727del2 | 5726_5727del	N/A	19	38979995	38979996

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Myopathy, congenital	RYR1	5749GtoT | E1917X	N/A	19	38980018	38980018
Malignant hyperthermia	RYR1	6349GtoC | V2117L	N/A	19	38985066	38985066
Malignant hyperthermia	RYR1	7372CtoT | R2458C	rs28933397	19	38991294	38991294
Malignant hyperthermia	RYR1	7373GtoA | R2458H	rs121918594	19	38991295	38991295
Malignant hyperthermia	RYR1	7373GtoT | R2458L	N/A	19	38991295	38991295
Nemaline myopathy	RYR1	7585GtoA | D2529N	N/A	19	38991601	38991601
Malignant hyperthermia, susceptibility to	RYR1	8327CtoT | S2776F	rs147707463	19	38995965	38995965
Myopathy, congenital	RYR1	8342_8343delTA | 8342_8343del2 | 8342_8343del	N/A	19	38995980	38995981
Malignant hyperthermia	RYR1	8353CtoG | L2785V	N/A	19	38995991	38995991
Multi-minicore disease	RYR1	8360CtoG | T2787S	rs35180584	19	38995998	38995998
Myopathy, congenital	RYR1	9579CtoG | C3193W	N/A	19	39006751	39006751
Malignant hyperthermia	RYR1	9649TtoC | S3217P	rs113422327	19	39006821	39006821
Myopathy, congenital	RYR1	10343CtoT | S3448F	N/A	19	39013751	39013751
Central core disease	RYR1	10616GtoA | R3539H	rs143987857	19	39016132	39016132
Central core disease	RYR1	12640_12648delCGCCAGTTC | 12640_12648del9 | 12640_12648del	rs118192165	19	39055614	39055622
Malignant hyperthermia, susceptibility to	RYR1	12960_12968dupGCGGCGGCT | 12960_12968dup9 | 12960_12968dup	N/A	19	39055942	39055943
Myopathy, congenital	RYR1	12986CtoA | A4329D	rs118192129	19	39055960	39055960
Myopathy, congenital (CNMDU1)	RYR1	13013_13032delCAGCAGTGACGCGCGCTGGG | 13013_13032del20 | 13013_13032del	N/A	19	39055987	39056006
Fibre type disproportion, congenital	RYR1	13480GtoT | E4494X	N/A	19	39057593	39057593
Malignant hyperthermia	RYR1	13502CtoT | P4501L	rs73933023	19	39057615	39057615
Myopathy, congenital	RYR1	13505AtoG | E4502G	rs139647387	19	39057618	39057618
Myopathy, congenital	RYR1	14173minus2AtoG	N/A	19	39068556	39068556
Glomerulosclerosis, focal and segmental	ACTN4	NULL	N/A	19	39138352	39138352
Spondylocostal dysostosis	DLL3	602delG | 602del1 | 602del	N/A	19	39993647	39993647
Spondylocostal dysostosis	DLL3	599_603dupGCGGT | 599_603dup5 | 599_603dup	N/A	19	39993648	39993649
Spondylocostal dysostosis	DLL3	1154GtoA | G385D	rs104894674	19	39997739	39997739
Otosclerosis	TGFB1	722CtoT | T241I	rs72480424	19	41847926	41847926
Osteoporosis, association with	TGFB1	713-8delC | 713-8del1 | 713-8del	N/A	19	41847943	41847943
Diamond-Blackfan anaemia	RPS19	320TtoG | L107R	N/A	19	42373248	42373248
Diamond-Blackfan anaemia	RPS19	328delC | 328del1 | 328del	N/A	19	42373256	42373256
Diamond-Blackfan anaemia	RPS19	340GtoT | E114X	N/A	19	42373268	42373268
Diamond-Blackfan anaemia	RPS19	344delA | 344del1 | 344del	N/A	19	42373272	42373272
Diamond-Blackfan anaemia	RPS19	343_344dupAA | 343_344dup2 | 343_344dup	N/A	19	42373272	42373273
Diamond-Blackfan anaemia	RPS19	356plus1GtoA	N/A	19	42373285	42373285
Diamond-Blackfan anaemia	RPS19	356plus1GtoT	N/A	19	42373285	42373285
Diamond-Blackfan anaemia	RPS19	356+1dupG | 356+1dup1 | 356+1dup	N/A	19	42373285	42373286
Diamond-Blackfan anaemia	RPS19	356plus2TtoA	N/A	19	42373286	42373286
Encephalopathy, ethylmalonic	ETHE1	604dupG | 604dup1 | 604dup	N/A	19	44012203	44012204
Hyperlipoproteinaemia	APOE	61GtoA | E21K	rs121918392	19	45411034	45411034
Apolipoprotein E deficiency	APOE	91GtoA | E31K	N/A	19	45411064	45411064
Hyperlipoproteinaemia III	APOE	114GtoA | W38X	N/A	19	45411087	45411087
Lipoprotein glomerulopathy	APOE	127CtoT | R43C	rs121918399	19	45411100	45411100
Alzheimer disease, increased risk	APOE	137TtoC | L46P	rs769452	19	45411110	45411110
Hyperlipoproteinaemia	APOE	146delG | 146del1 | 146del	N/A	19	45411119	45411119
Hyperlipidaemia	APOE	192GtoC | Q64H	N/A	19	45411165	45411165
Apolipoprotein E deficiency	APOE	340dupG | 340dup1 | 340dup	N/A	19	45411893	45411894
Hermansky-Pudlak syndrome	BLOC1S3	131CtoA | S44X	N/A	19	45682685	45682685
Xeroderma pigmentosum	ERCC2	1745_1747delAGAinsTTTCGG | 1745_1747del3ins6 | 1745_1747delins	N/A	19	45856511	45856513
Xeroderma pigmentosum	ERCC2	595minus10GtoA	N/A	19	45867815	45867815
Spastic paraplegia 12	RTN2	881CtoT | S294F	N/A	19	45992745	45992745
Muscular dystrophy, limb girdle 2I	FKRP	1030GtoT | G344C	N/A	19	47259737	47259737
Reduced activity	SULT2B1	689GtoA | R230H	rs16982169	19	49100039	49100039
Infertility, female, association with	LHB	364GtoA | G122S	rs5030774	19	49519387	49519387
Heart block type 1	TRPM4	19GtoA | E7K	N/A	19	49661142	49661142
Cataract	LIM2	439TtoG | F147V	rs121913555	19	51885684	51885684
Spinocerebellar ataxia 14	PRKCG	1927TtoC | F643L	rs121918516	19	54409982	54409982
Retinitis pigmentosa	PRKCG	1975CtoA | R659S	rs139735546	19	54410030	54410030
Retinitis pigmentosa	PRPF31	973GtoT | E325X	N/A	19	54631475	54631475
Retinitis pigmentosa	PRPF31	997delG | 997del1 | 997del	N/A	19	54631499	54631499
Retinitis pigmentosa	PRPF31	1115_1125delGGAAGCAGGCC | 1115_1125del11 | 1115_1125del	N/A	19	54631721	54631731
Retinitis pigmentosa	PRPF31	1142delG | 1142del1 | 1142del	N/A	19	54631748	54631748
Retinitis pigmentosa	PRPF31	1146plus2TtoC	N/A	19	54631754	54631754
Retinitis pigmentosa	PRPF31	1155_1159delGGACGinsAGGGATT | 1155_1159del5ins7 | 1155_1159delins	N/A	19	54632440	54632444
Cardiomyopathy, hypertrophic	TNNI3	61CtoT | R21C	N/A	19	55668465	55668465
Large-headed spermatazoa	AURKC	436minus2AtoG	N/A	19	57744826	57744826
Diabetes insipidus, neurohypophyseal	AVP	352GtoT | E118X	N/A	20	3063419	3063419
Diabetes insipidus, neurohypophyseal	AVP	348CtoG | C116W	N/A	20	3063423	3063423
Diabetes insipidus, neurohypophyseal	AVP	346TtoC | C116R	N/A	20	3063425	3063425
Diabetes insipidus, neurohypophyseal	AVP	346TtoG | C116G	rs74315383	20	3063425	3063425
Diabetes insipidus, central	AVP	343GtoT | E115X	N/A	20	3063428	3063428
Diabetes insipidus, neurohypophyseal	AVP	342_343delCGinsGT | 342_343del2ins2 | 342_343delins	N/A	20	3063428	3063429
Diabetes insipidus, neurohypophyseal	AVP	337GtoT | E113X	rs121964889	20	3063434	3063434
Diabetes insipidus, neurohypophyseal	AVP	330CtoA | C110X	N/A	20	3063441	3063441
Corneal endothelial dystrophy 2	SLC4A11	2066plus1GtoA	N/A	20	3209740	3209740
Corneal endothelial dystrophy 2	SLC4A11	2017_2019delTTC | 2017_2019del3 | 2017_2019del	N/A	20	3209788	3209790
Kindler syndrome	FERMT1	614GtoA | W205X	N/A	20	6091077	6091077
Bone mass, association with	BMP2	109TtoG | S37A	rs2273073	20	6750882	6750882
Alagille syndrome	JAG1	1964delG | 1964del1 | 1964del	N/A	20	10626654	10626654
Alagille syndrome	JAG1	852_853delTG | 852_853del2 | 852_853del	N/A	20	10633149	10633150
Alagille syndrome	JAG1	851GtoT | C284F	N/A	20	10633151	10633151
Alagille syndrome	JAG1	841CtoT | Q281X	N/A	20	10633161	10633161
Alagille syndrome	JAG1	839GtoA | W280X	N/A	20	10633163	10633163
Tetralogy of Fallot	JAG1	834GtoT | E278D	N/A	20	10633168	10633168
Alagille syndrome	JAG1	830delA | 830del1 | 830del	N/A	20	10633172	10633172
Alagille syndrome	JAG1	826delT | 826del1 | 826del	N/A	20	10633176	10633176
Alagille syndrome	JAG1	794GtoT | C265F	N/A	20	10633208	10633208
Alagille syndrome	JAG1	776GtoT | G259V	N/A	20	10633226	10633226
Alagille syndrome	JAG1	770GtoA | W257X	N/A	20	10633232	10633232
Alagille syndrome	JAG1	756_762delGTGCCAG | 756_762del7 | 756_762del	N/A	20	10633240	10633246
Alagille syndrome	JAG1	66_67delTG | 66_67del2 | 66_67del	N/A	20	10654112	10654113
Alagille syndrome	JAG1	64TtoC | C22R	N/A	20	10654115	10654115
Alagille syndrome	JAG1	59TtoC | L20P	N/A	20	10654120	10654120
Alagille syndrome	JAG1	41delT | 41del1 | 41del	N/A	20	10654138	10654138
Alagille syndrome	JAG1	40delC | 40del1 | 40del	N/A	20	10654139	10654139
Alagille syndrome	JAG1	30_36delCGGGCGCinsACTCGGGA | 30_36del7ins8 | 30_36delins	N/A	20	10654143	10654149

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Anaemia, dyserythropoietic congenital, type II	SEC23B	2150delC | 2150del1 | 2150del	N/A	20	18535753	18535753
Inner retina abnormality	VSX1	740_741delCGinsGA | 740_741del2ins2 | 740_741delins	N/A	20	25058388	25058389
Keratoconus	VSX1	740CtoG | P247R	N/A	20	25058389	25058389
Keratoconus	VSX1	731AtoG | H244R	rs148957473	20	25058398	25058398
Keratoconus	VSX1	715GtoC | G239R	N/A	20	25058414	25058414
Posterior polymorphous dystrophy	VSX1	479GtoA | G160D	rs74315433	20	25060096	25060096
Keratoconus	VSX1	479GtoT | G160V	rs74315433	20	25060096	25060096
Keratoconus	VSX1	475TtoA | L159M	rs74315434	20	25060100	25060100
Keratoconus	VSX1	452AtoG | N151S	N/A	20	25060123	25060123
Posterior polymorphous dystrophy	VSX1	432CtoG | D144E	rs140122268	20	25060143	25060143
Polyneuropathy, hearing loss, ataxia, retinitis p	ABHD12	1054CtoT | R352X	N/A	20	25282958	25282958
Reduced activity	AHCY	367GtoA | G123R	rs41301825	20	32880242	32880242
Glutathione synthetase deficiency	GSS	809AtoG | Y270C	N/A	20	33523404	33523404
Glutathione synthetase deficiency	GSS	808TtoC | Y270H	N/A	20	33523405	33523405
Glutathione synthetase deficiency	GSS	799CtoT | R267W	rs121909308	20	33523414	33523414
Glutathione synthetase deficiency	GSS	799CtoG	N/A	20	33523414	33523414
Reduced activity	TGM2	641GtoA | R214H	rs45530133	20	36776403	36776403
Cardiomyopathy, hypertrophic	JPH2	494CtoT | S165F	N/A	20	42788933	42788933
Diabetes, MODY	HNF4A	1193TtoC | M398T	N/A	20	43057038	43057038
Diabetes, MODY1	HNF4A	1204GtoA | V402I	rs137853337	20	43057049	43057049
Hypercalcaemia, idiopathic infantile	CYP24A1	476GtoA | R159Q	N/A	20	52788183	52788183
Hypercalcaemia, idiopathic infantile	CYP24A1	451GtoT | E151X	N/A	20	52788208	52788208
Albright hereditary osteodystrophy	GNAS	1AtoG | M1V	rs137854530	20	57466782	57466782
Albright hereditary osteodystrophy	GNAS	2TtoG | M1R	N/A	20	57466783	57466783
Albright hereditary osteodystrophy	GNAS	3GtoA | M1I	N/A	20	57466784	57466784
Albright hereditary osteodystrophy	GNAS	21dupT | 21dup1 | 21dup	N/A	20	57466802	57466803
Progressive osseous heteroplasia	GNAS	34CtoT | Q12X	N/A	20	57466815	57466815
Pseudohypoparathyroidism 1a	GNAS	70_74delAAAAAinsTGCGCTGCAGAAG | 70_74del5ins13 | 70_74delins	N/A	20	57466851	57466855
Albright hereditary osteodystrophy	GNAS	77TtoA | I26N	N/A	20	57466858	57466858
Pseudohypoparathyroidism 1a	GNAS	83dupA | 83dup1 | 83dup	N/A	20	57466864	57466865
Progressive osseous heteroplasia	GNAS	85CtoT | Q29X	N/A	20	57466866	57466866
Albright hereditary osteodystrophy	GNAS	91CtoT | Q31X	N/A	20	57466872	57466872
Albright hereditary osteodystrophy	GNAS	93delG | 93del1 | 93del	N/A	20	57466874	57466874
Pseudohypoparathyroidism 1a	GNAS	94AtoT | K32X	N/A	20	57466875	57466875
Pseudohypoparathyroidism 1a	GNAS	103CtoT | Q35X	N/A	20	57466884	57466884
Albright hereditary osteodystrophy	GNAS	111CtoG | Y37X	N/A	20	57466892	57466892
Pseudohypoparathyroidism 1a	GNAS	112delC | 112del1 | 112del	N/A	20	57466893	57466893
Albright hereditary osteodystrophy	GNAS	115delG | 115del1 | 115del	N/A	20	57466896	57466896
Waardenburg syndrome 4	EDN3	277CtoG | R93G	N/A	20	57876689	57876689
Shah-Waardenburg syndrome	EDN3	286CtoT | R96C	N/A	20	57876698	57876698
Shah-Waardenburg syndrome	EDN3	293CtoA | T98K	N/A	20	57876705	57876705
Waardenburg syndrome 4B	EDN3	293CtoT | T98M	N/A	20	57876705	57876705
Epilepsy, benign neonatal	KCNQ2	684CtoA | H228Q	rs118192204	20	62076018	62076018
Epilepsy, benign neonatal	KCNQ2	640CtoT | R214W	rs28939684	20	62076062	62076062
Epileptic encephalopathy, neonatal	KCNQ2	638GtoA | R213Q	N/A	20	62076064	62076064
Epilepsy, benign neonatal	KCNQ2	637CtoT | R213W	rs118192203	20	62076065	62076065
Epilepsy, benign neonatal	KCNQ2	635AtoG | D212G	rs118192202	20	62076067	62076067
Epilepsy, benign neonatal	KCNQ2	622AtoG | M208V	rs118192201	20	62076080	62076080
Peripheral nerve hyperexcitability	KCNQ2	620GtoA | R207Q	rs118192200	20	62076082	62076082
Epilepsy, benign neonatal	KCNQ2	619CtoT | R207W	rs74315391	20	62076083	62076083
Epileptic encephalopathy, neonatal	KCNQ2	613AtoG | I205V	N/A	20	62076089	62076089
Epilepsy, benign neonatal	KCNQ2	584_593delCTGCGCTCCGinsA | 584_593del10ins1 | 584_593delins	N/A	20	62076109	62076118
Epilepsy, benign neonatal	KCNQ2	587_590delCGCTinsTGCC | 587_590del4ins4 | 587_590delins	N/A	20	62076112	62076115
Epilepsy, benign neonatal	KCNQ2	587CtoT | A196V	rs118192199	20	62076115	62076115
Epilepsy, benign neonatal	KCNQ2	585dupT | 585dup1 | 585dup	N/A	20	62076116	62076117
Platelet disorder, familial	RUNX1	735delC | 735del1 | 735del	N/A	21	36206777	36206777
Bartsocas-Papas syndrome	RIPK4	777dupA | 777dup1 | 777dup	N/A	21	43166827	43166828
Bartsocas-Papas syndrome	RIPK4	362TtoA | I121N	N/A	21	43176797	43176797
Homocystinuria	CBS	727CtoT | Q243X	N/A	21	44485322	44485322
Epilepsy, progressive myoclonus	CSTB	66GtoA	N/A	21	45196085	45196085
APECED	AIRE	22CtoT | R8C	N/A	21	45705911	45705911
APECED	AIRE	43CtoT | R15C	rs179363875	21	45705932	45705932
APECED	AIRE	44GtoT | R15L	rs179363876	21	45705933	45705933
APECED	AIRE	47CtoT | T16M	rs179363877	21	45705936	45705936
APECED	AIRE	55GtoA | A19T	N/A	21	45705944	45705944
APECED	AIRE	62CtoT | A21V	rs179363886	21	45705951	45705951
APECED	AIRE	64_69delGTGGAC | 64_69del6 | 64_69del	N/A	21	45705953	45705958
APECED	AIRE	892GtoA | E298K	N/A	21	45710990	45710990
Addison disease with autoimmune thyroiditis	AIRE	901GtoA | V301M	rs150634562	21	45710999	45710999
APECED	AIRE	905_906delGT | 905_906del2 | 905_906del	N/A	21	45711003	45711004
APECED	AIRE	906TtoA | C302X	N/A	21	45711004	45711004
APECED	AIRE	908GtoC | R303P	N/A	21	45711006	45711006
Autoimmune hepatitis 2	AIRE	913GtoA | G305S	N/A	21	45711011	45711011
APECED	AIRE	1096minus1GtoA	N/A	21	45712875	45712875
APECED	AIRE	1096minus1GtoC	N/A	21	45712875	45712875
APECED	AIRE	1103dupC | 1103dup1 | 1103dup	N/A	21	45712883	45712884
Hypoparathyroidism without APECED	AIRE	1155dupA | 1155dup1 | 1155dup	N/A	21	45712935	45712936
APECED	AIRE	1163_1164insA | 1163_1164ins1 | 1163_1164ins	N/A	21	45712943	45712944
APECED	AIRE	1189delC | 1189del1 | 1189del	N/A	21	45712969	45712969
APECED	AIRE	1193delC | 1193del1 | 1193del	N/A	21	45712973	45712973
APECED	AIRE	1236_1239dupGGCC | 1236_1239dup4 | 1236_1239dup	N/A	21	45713019	45713020
APECED	AIRE	1242_1243insA | 1242_1243ins1 | 1242_1243ins	N/A	21	45713022	45713023
APECED	AIRE	1249delC | 1249del1 | 1249del	N/A	21	45713029	45713029
APECED	AIRE	1249dupC | 1249dup1 | 1249dup	N/A	21	45713029	45713030
APECED	AIRE	1265delC | 1265del1 | 1265del	N/A	21	45713045	45713045
Leukocyte adhesion deficiency	ITGB2	130AtoC | T44P	N/A	21	46330216	46330216
Leukocyte adhesion deficiency	ITGB2	119_128delGGCCCGGCTG | 119_128del10 | 119_128del	N/A	21	46330218	46330227
Leukocyte adhesion deficiency	ITGB2	120delG | 120del1 | 120del	N/A	21	46330226	46330226
Leukocyte adhesion deficiency	ITGB2	106TtoA | C36S	N/A	21	46330240	46330240
Leukocyte adhesion deficiency	ITGB2	79AtoT | K27X	rs189250711	21	46330267	46330267
Leukocyte adhesion deficiency	ITGB2	77dupC | 77dup1 | 77dup	N/A	21	46330268	46330269
Leukocyte adhesion deficiency	ITGB2	66_67delTC | 66_67del2 | 66_67del	N/A	21	46330279	46330280
Leukocyte adhesion deficiency	ITGB2	59minus10CtoA	N/A	21	46330297	46330297
Bethlem myopathy	COL6A1	1003minus3CtoA	N/A	21	47410684	47410684
Ullrich congenital muscular dystrophy	COL6A2	1096CtoT | R366X	N/A	21	47537830	47537830

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Ullrich congenital muscular dystrophy	COL6A2	1116plus1GtoT	N/A	21	47537851	47537851
Ullrich congenital muscular dystrophy	COL6A2	1270minus1GtoC	N/A	21	47539701	47539701
Ullrich congenital muscular dystrophy	COL6A2	1591GtoC | G531R	N/A	21	47542428	47542428
Glutamate formiminotransferase deficiency	FTCD	990dupG | 990dup1 | 990dup	N/A	21	47565839	47565840
Glutamate formiminotransferase deficiency	FTCD	403CtoT | R135C	rs28941768	21	47571859	47571859
Hyperprolinaemia, association with	PRODH	1414GtoA | A472T	rs2870983	22	18905842	18905842
Hyperprolinaemia, association with	PRODH	1397CtoT | T466M	rs2870984	22	18905859	18905859
Increased proline level in schizophrenia	PRODH	1363GtoT | A455S	rs1807467	22	18905893	18905893
Hyperprolinaemia, association with	PRODH	1357CtoT | R453C	rs3970559	22	18905899	18905899
Hyperprolinaemia, association with	PRODH	1331GtoA | G444D	N/A	22	18905925	18905925
Increased proline level in schizophrenia	PRODH	1322TtoC | L441P	rs2904551	22	18905934	18905934
Hyperprolinaemia, association with	PRODH	172GtoA | A58T	rs146648839	22	18923629	18923629
Hyperprolinaemia, association with	PRODH	88CtoT | P30S	rs3815655	22	18923713	18923713
Bernard-Soulier syndrome	GP1BB	89GtoA | C30Y	N/A	22	19711455	19711455
Giant platelet disorder	GP1BB	124CtoT | R42C	N/A	22	19711490	19711490
Giant platelet disorder	GP1BB	137GtoA | W46X	rs121909752	22	19711503	19711503
Bernard-Soulier syndrome	GP1BB	143CtoA | S48X	N/A	22	19711509	19711509
Bernard-Soulier syndrome	GP1BB	266AtoC | N89T	N/A	22	19711632	19711632
Bernard-Soulier syndrome	GP1BB	269CtoG | P90R	N/A	22	19711635	19711635
Giant platelet disorder	GP1BB	296CtoG | P99R	N/A	22	19711662	19711662
Cardiovascular defects	TBX1	1187CtoT | P396L	N/A	22	19754089	19754089
Cardiomyopathy, dilated	TXNRD2	1123GtoA | G375R	N/A	22	19868204	19868204
Bipolar disorder, association with	BCR	2387AtoG | N796S	rs140504	22	23627369	23627369
Increased expression	SMARCB1	NULL	rs11704810	22	24129129	24129129
Kleefstra syndrome	SMARCB1	110GtoA | R37H	N/A	22	24133959	24133959
Rhabdoid tumour	SMARCB1	118CtoT | R40X	N/A	22	24133967	24133967
Rhabdoid tumour	SMARCB1	122dupG | 122dup1 | 122dup	N/A	22	24133971	24133972
Breast cancer	CHEK2	846+4_846+7delAGTA | 846+4_846+7del4 | 846+4_846+7del	N/A	22	29105987	29105990
Breast cancer	CHEK2	825_826delAA | 825_826del2 | 825_826del	N/A	22	29106014	29106015
Breast cancer	CHEK2	823GtoT | E275X	N/A	22	29106017	29106017
Complex I deficiency	NIPSNAP1	215AtoG | Y72C	N/A	22	29966405	29966405
Neurofibromatosis 2	NF2	364minus1GtoA	N/A	22	30038190	30038190
Neurofibromatosis 2	NF2	364minus1GtoT	N/A	22	30038190	30038190
Neurofibromatosis 2	NF2	367AtoT | K123X	N/A	22	30038194	30038194
Neurofibromatosis 2	NF2	397TtoC | C133R	N/A	22	30038224	30038224
Neurofibromatosis 2	NF2	415_429delGTGCTCCTGGCTTCT | 415_429del15 | 415_429del	N/A	22	30038242	30038256
Neurofibromatosis 2	NF2	422TtoC | L141P	N/A	22	30038249	30038249
Neurofibromatosis 2	NF2	420_426dupCCTGGCT | 420_426dup7 | 420_426dup	N/A	22	30038253	30038254
Neurofibromatosis 2	NF2	431_432insGA | 431_432ins2 | 431_432ins	N/A	22	30038258	30038259
Neurofibromatosis 2	NF2	431dupA | 431dup1 | 431dup	N/A	22	30038258	30038259
Neurofibromatosis 2	NF2	432CtoA | Y144X	N/A	22	30038259	30038259
Neurofibromatosis 2	NF2	432CtoG | Y144X	N/A	22	30038259	30038259
Neurofibromatosis 2	NF2	448minus2AtoG	N/A	22	30050644	30050644
Neurofibromatosis 2	NF2	448minus2AtoT	N/A	22	30050644	30050644
Neurofibromatosis 2	NF2	448minus1GtoA	N/A	22	30050645	30050645
Neurofibromatosis 2	NF2	448minus1GtoT	N/A	22	30050645	30050645
Neurofibromatosis 2	NF2	459CtoA | Y153X	N/A	22	30050657	30050657
Neurofibromatosis 2	NF2	459CtoG | Y153X	N/A	22	30050657	30050657
Neurofibromatosis 2	NF2	463_464delCCinsT | 463_464del2ins1 | 463_464delins	N/A	22	30050661	30050662
Neurofibromatosis 2	NF2	461_479dupACCCCAGTGTTCACAAGCG | 461_479dup19 | 461_479dup	N/A	22	30050677	30050678
Neurofibromatosis 2	NF2	482delG | 482del1 | 482del	N/A	22	30050680	30050680
Neurofibromatosis 2	NF2	493CtoT | Q165X	N/A	22	30050691	30050691
Neurofibromatosis 2	NF2	1018delG | 1018del1 | 1018del	N/A	22	30067833	30067833
Neurofibromatosis 2	NF2	1021CtoT | R341X	rs74315499	22	30067836	30067836
Neurofibromatosis 2	NF2	1408CtoT | Q470X	N/A	22	30070892	30070892
Neurofibromatosis 2	NF2	1417delC | 1417del1 | 1417del	N/A	22	30070901	30070901
Iron-refractory iron deficiency anaemia (IRIDA	TMPRSS6	1682CtoA | S561X	rs143441292	22	37466973	37466973
Iron-refractory iron deficiency anaemia (IRIDA	TMPRSS6	1669TtoA | C557S	N/A	22	37466986	37466986
Iron-refractory iron deficiency anaemia (IRIDA	TMPRSS6	1065CtoA | Y355X	rs137853121	22	37480815	37480815
Iron-refractory iron deficiency anaemia (IRIDA	TMPRSS6	658plus1GtoC	N/A	22	37491590	37491590
Iron-refractory iron deficiency anaemia (IRIDA	TMPRSS6	635TtoC | I212T	N/A	22	37491614	37491614
Infantile neuroaxonal dystrophy 1	PLA2G6	1538_1541delACTG | 1538_1541del4 | 1538_1541del	N/A	22	38519152	38519155
Hypercholesterolaemia	SREBF2	1934GtoA | R645Q	N/A	22	42276892	42276892
N-acetylgalactosaminidase alpha deficiency	NAGA	577GtoT | E193X	rs121434531	22	42462734	42462734
Diabetes	PPARA	1226GtoC | R409T	rs1800243	22	46631096	46631096
Craniorachischisis	CELSR1	7313GtoA | R2438Q	N/A	22	46774558	46774558
Ulcerative colitis, increased risk, association w	IL17REL	998TtoC | L333P	rs5771069	22	50435480	50435480
Megalencephalic leukoencephalopathy	MLC1	927_929dupGCT | 927_929dup3 | 927_929dup	N/A	22	50502592	50502593
Megalencephalic leukoencephalopathy	MLC1	924_929delGCTGCT | 924_929del6 | 924_929del	N/A	22	50502593	50502598
Catatonic schizophrenia	MLC1	925CtoA | L309M	rs80358240	22	50502597	50502597
Bipolar affective disorder	MLC1	923TtoA | L308Q	N/A	22	50502599	50502599
Megalencephalic leukoencephalopathy	MLC1	908_918delTGCTGCTGCTGinsGCA | 908_918del11ins3 | 908_918delins	N/A	22	50502604	50502614
Megalencephalic leukoencephalopathy	MLC1	895minus1GtoC	N/A	22	50502628	50502628
Megalencephalic leukoencephalopathy	MLC1	895minus2AtoG	N/A	22	50502629	50502629
Megalencephalic leukoencephalopathy with s	MLC1	772minus1GtoC	N/A	22	50506985	50506985
Megalencephalic leukoencephalopathy	MLC1	597plus1GtoA	N/A	22	50515269	50515269
Megalencephalic leukoencephalopathy	MLC1	594_597delCTCA | 594_597del4 | 594_597del	N/A	22	50515270	50515273
Mitochondrial neurogastrointestinal encephal	TYMP	1311delG | 1311del1 | 1311del	N/A	22	50964337	50964337
Mitochondrial neurogastrointestinal encephal	TYMP	1310GtoA | W437X	N/A	22	50964338	50964338
Mitochondrial neurogastrointestinal encephal	TYMP	1301minus1GtoA	N/A	22	50964348	50964348
Mitochondrial neurogastrointestinal encephal	TYMP	1211dupT | 1211dup1 | 1211dup	N/A	22	50964518	50964519
Mitochondrial neurogastrointestinal encephal	TYMP	931GtoC | G311R	rs121913040	22	50964903	50964903
Mitochondrial neurogastrointestinal encephal	TYMP	931GtoT | G311C	N/A	22	50964903	50964903
Arylsulphatase A pseudodeficiency, associatio	ARSA	1055AtoG | N352S	rs2071421	22	51064416	51064416
Metachromatic leukodystrophy	ARSA	1046delC | 1046del1 | 1046del	N/A	22	51064425	51064425
Autism spectrum disorder	SHANK3	34CtoT | R12C	N/A	22	51113103	51113103
Intellectual disability, nonsyndromic	SHANK3	601minus1GtoA	N/A	22	51117446	51117446
Schizophrenia	SHANK3	1654CtoT | R552W	N/A	22	51137225	51137225
Chondrodysplasia punctata, X-linked	ARSE	119TtoG | I40S	rs80338710	X	2876381	2876381
Kallmann syndrome	KAL1	1720dupG | 1720dup1 | 1720dup	N/A	X	8503753	8503754
Kallmann syndrome	KAL1	3GtoA | M1I	N/A	X	8700075	8700075
Albinism, ocular	GPR143	180dupC | 180dup1 | 180dup	N/A	X	9733677	9733678
Albinism, ocular	GPR143	175delT | 175del1 | 175del	rs62635023	X	9733683	9733683
Albinism, ocular	GPR143	163_170dupGCGGGCCC | 163_170dup8 | 163_170dup	N/A	X	9733687	9733688
Albinism, ocular	GPR143	155_161delGCCGGCC | 155_161del7 | 155_161del	N/A	X	9733697	9733703

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Albinism, ocular	GPR143	116TtoG | L39R	rs62635019	X	9733742	9733742
Nance-Horan syndrome	NHS	115CtoT | Q39X	rs104894881	X	17393995	17393995
Nance-Horan syndrome	NHS	792delA | 792del1 | 792del	N/A	X	17710528	17710528
Encephalopathy with early epilepsy	CDKL5	2066delC | 2066del1 | 2066del	N/A	X	18627604	18627604
Mental disability, hypotonia, and intractable e	CDKL5	2105_2106delAC | 2105_2106del2 | 2105_2106del	N/A	X	18627643	18627644
Mental retardation and epilepsy	CDKL5	2376plus1GtoA	N/A	X	18638087	18638087
Rett syndrome, variant, with infantile spasms	CDKL5	2376plus1GtoC	N/A	X	18638087	18638087
Retinoschisis, X linked juvenile	RS1	276GtoC | W92C	rs61752062	X	18665361	18665361
Retinoschisis, X linked	RS1	274TtoG | W92G	N/A	X	18665363	18665363
Retinoschisis, X linked juvenile	RS1	267TtoA | Y89X	rs61752061	X	18665370	18665370
Retinoschisis, X linked juvenile	RS1	266AtoG | Y89C	rs61752060	X	18665371	18665371
Retinoschisis, X linked juvenile	RS1	262CtoT | Q88X	rs61750459	X	18665375	18665375
Retinoschisis, X linked juvenile	RS1	253_255delAAC | 253_255del3 | 253_255del	rs61750458	X	18665382	18665384
Retinoschisis, X linked juvenile	RS1	242TtoA | I81N	rs61750457	X	18665395	18665395
Lactic acidosis	PDHA1	844AtoC | M282L	rs2229137	X	19375782	19375782
Pyruvate dehydrogenase deficiency	PDHA1	844AtoG | M282V	N/A	X	19375782	19375782
Potential protein deficiency	MAP3K15	3364CtoT | R1122X	rs140104197	X	19389113	19389113
Coffin-Lowry syndrome	RPS6KA3	69+1delG | 69+1del1 | 69+1del	N/A	X	20284681	20284681
X-chromosomal hearing loss	SMPX	109GtoT | E37X	N/A	X	21761891	21761891
Lissencephaly, X-linked, with abnormal genital	ARX	232GtoT | E78X	N/A	X	25031880	25031880
Muscular dystrophy, Becker	DMD	10910CtoA | S3637X	N/A	X	31164419	31164419
Muscular dystrophy, Duchenne	DMD	10903CtoT | Q3635X	N/A	X	31164426	31164426
Muscular dystrophy, Becker	DMD	10888CtoT | R3630X	N/A	X	31164441	31164441
Muscular dystrophy, Becker	DMD	10812delC | 10812del1 | 10812del	N/A	X	31164517	31164517
Muscular dystrophy, Duchenne	DMD	10801CtoT | Q3601X	N/A	X	31164528	31164528
Muscular dystrophy, Becker	DMD	9650-2_9654delAGACCTT | 9650-2_9654del7 | 9650-2_9654del	N/A	X	31222231	31222237
Muscular dystrophy, Duchenne	DMD	9224plus5GtoC	N/A	X	31341710	31341710
Muscular dystrophy, Duchenne	DMD	9224plus1GtoA	N/A	X	31341714	31341714
Muscular dystrophy, Duchenne	DMD	9224plus1GtoC	N/A	X	31341714	31341714
Muscular dystrophy, Duchenne	DMD	9224plus1GtoT	N/A	X	31341714	31341714
Chronic granulomatous disease	CYBB	175TtoC | C59R	rs151344457	X	37642776	37642776
Chronic granulomatous disease	CYBB	176GtoA | C59Y	N/A	X	37642777	37642777
Chronic granulomatous disease	CYBB	176GtoT | C59F	N/A	X	37642777	37642777
Chronic granulomatous disease	CYBB	177CtoA | C59X	N/A	X	37642778	37642778
Chronic granulomatous disease	CYBB	177CtoG | C59W	rs151344488	X	37642778	37642778
Chronic granulomatous disease	CYBB	185delT | 185del1 | 185del	N/A	X	37642786	37642786
Chronic granulomatous disease	CYBB	189CtoG | N63K	N/A	X	37642790	37642790
Chronic granulomatous disease	CYBB	190TtoC | C64R	N/A	X	37642791	37642791
Chronic granulomatous disease	CYBB	192CtoA | C64X	N/A	X	37642793	37642793
Chronic granulomatous disease	CYBB	194TtoG | M65R	N/A	X	37642795	37642795
Chronic granulomatous disease	CYBB	195dupG | 195dup1 | 195dup	N/A	X	37642796	37642797
Chronic granulomatous disease	CYBB	197TtoC | L66P	N/A	X	37642798	37642798
Chronic granulomatous disease	CYBB	210dupA | 210dup1 | 210dup	N/A	X	37642811	37642812
Chronic granulomatous disease	CYBB	217CtoT | R73X	rs137854588	X	37642818	37642818
Chronic granulomatous disease	CYBB	226delC | 226del1 | 226del	N/A	X	37642827	37642827
Chronic granulomatous disease	CYBB	548_559delCGCTGTGCCTCA | 548_559del12 | 548_559del	N/A	X	37655268	37655279
Chronic granulomatous disease	CYBB	553TtoC | C185R	N/A	X	37655273	37655273
Chronic granulomatous disease	CYBB	554delG | 554del1 | 554del	N/A	X	37655274	37655274
Chronic granulomatous disease	CYBB	1151plus5GtoA	N/A	X	37663388	37663388
Mental retardation & microcephaly with ponti	CASK	2632CtoT | Q878X	N/A	X	41379807	41379807
Norrie disease	NDP	174plus1GtoC	N/A	X	43817717	43817717
Norrie disease	NDP	174GtoT | K58N	N/A	X	43817718	43817718
Norrie disease	NDP	170CtoG | S57X	N/A	X	43817722	43817722
Norrie disease	NDP	164GtoT | C55F	N/A	X	43817728	43817728
Norrie disease	NDP	163TtoC | C55R	N/A	X	43817729	43817729
Norrie disease	NDP	162GtoC | K54N	N/A	X	43817730	43817730
Norrie disease	NDP	142_145delATCA | 142_145del4 | 142_145del	N/A	X	43817747	43817750
Norrie disease	NDP	136delG | 136del1 | 136del	N/A	X	43817756	43817756
Norrie disease	NDP	134TtoA | V45E	rs137852221	X	43817758	43817758
Norrie disease	NDP	133GtoA | V45M	N/A	X	43817759	43817759
Norrie disease	NDP	131dupA | 131dup1 | 131dup	N/A	X	43817760	43817761
Norrie disease	NDP	131AtoG | Y44C	rs104894870	X	43817761	43817761
Norrie disease	NDP	129CtoG | H43Q	N/A	X	43817763	43817763
Norrie disease	NDP	129delC | 129del1 | 129del	N/A	X	43817763	43817763
Norrie disease	NDP	128dupA | 128dup1 | 128dup	N/A	X	43817763	43817764
Norrie disease	NDP	128AtoG | H43R	N/A	X	43817764	43817764
Norrie disease	NDP	125AtoG | H42R	rs104894874	X	43817767	43817767
Retinal malformation	NDP	123GtoC | R41S	N/A	X	43817769	43817769
Bilateral persistant fetal vasculature	NDP	123GtoT | R41S	N/A	X	43817769	43817769
Norrie disease	NDP	122GtoA | R41K	N/A	X	43817770	43817770
Norrie disease	NDP	122GtoC | R41T	N/A	X	43817770	43817770
Retinal malformation	NDP	115TtoC | C39R	N/A	X	43817777	43817777
Norrie disease	NDP	112CtoT | R38C	N/A	X	43817780	43817780
Norrie disease	NDP	109CtoT | R37X	N/A	X	43817783	43817783
Norrie disease	NDP	86CtoG | S29X	N/A	X	43817806	43817806
Norrie disease	NDP	65delC | 65del1 | 65del	N/A	X	43817827	43817827
Exudative vitreoretinopathy	NDP	53TtoA | I18K	N/A	X	43817839	43817839
Norrie disease	NDP	50dupT | 50dup1 | 50dup	N/A	X	43817841	43817842
Norrie disease	NDP	49delG | 49del1 | 49del	N/A	X	43817843	43817843
Norrie disease	NDP	47TtoC | L16P	N/A	X	43817845	43817845
Norrie disease	NDP	25_40delTCCTTTTCTATGCTCT | 25_40del16 | 25_40del	N/A	X	43817852	43817867
Norrie disease	NDP	38TtoG | L13R	rs104894879	X	43817854	43817854
Norrie disease	NDP	24_27dupATCC | 24_27dup4 | 24_27dup	N/A	X	43817864	43817865
Autism / epilepsy	SYN1	1648GtoA | A550T	N/A	X	47433735	47433735
Focal dermal hypoplasia	PORCN	1344GtoA | W448X	N/A	X	48378822	48378822
Focal dermal hypoplasia	PORCN	1353dupT | 1353dup1 | 1353dup	N/A	X	48378831	48378832
Focal dermal hypoplasia	PORCN	1361GtoA | W454X	N/A	X	48378839	48378839
Chondrodysplasia punctata, X-linked	EBP	523CtoT | Q175X	rs104894793	X	48386675	48386675
Chondrodysplasia punctata, X-linked	EBP	540_541delCC | 540_541del2 | 540_541del	N/A	X	48386692	48386693
Chondrodysplasia punctata, X-linked	EBP	552CtoG | Y184X	N/A	X	48386704	48386704
Chondrodysplasia punctata, X-linked	EBP	558GtoA | W186X	N/A	X	48386710	48386710
Chondrodysplasia punctata, X-linked	EBP	608TtoC | L203P	N/A	X	48386760	48386760
Conradi-Hunermann-Happle syndrome	EBP	616GtoT | D206Y	N/A	X	48386768	48386768
Wiskott-Aldrich syndrome	WAS	607CtoT | Q203X	N/A	X	48545217	48545217
Wiskott-Aldrich syndrome	WAS	611delA | 611del1 | 611del	N/A	X	48545221	48545221

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Wiskott-Aldrich syndrome	WAS	628delT | 628del1 | 628del	N/A	X	48545238	48545238
Wiskott-Aldrich syndrome	WAS	631CtoT | R211X	N/A	X	48545241	48545241
Wiskott-Aldrich syndrome	WAS	647dupC | 647dup1 | 647dup	N/A	X	48545257	48545258
Wiskott-Aldrich syndrome	WAS	649_652dupGCAC | 649_652dup4 | 649_652dup	N/A	X	48545262	48545263
Wiskott-Aldrich syndrome	WAS	647_659dupCAGCACCTGGACC | 647_659dup13 | 647_659dup	N/A	X	48545269	48545270
Wiskott-Aldrich syndrome	WAS	661dupA | 661dup1 | 661dup	N/A	X	48545271	48545272
Wiskott-Aldrich syndrome	WAS	671AtoG | D224G	N/A	X	48545281	48545281
Wiskott-Aldrich syndrome	WAS	671AtoG	N/A	X	48545281	48545281
Wiskott-Aldrich syndrome	WAS	683CtoG | S228X	N/A	X	48545293	48545293
Wiskott-Aldrich syndrome	WAS	687GtoT	N/A	X	48545297	48545297
Wiskott-Aldrich syndrome	WAS	697delA | 697del1 | 697del	N/A	X	48545307	48545307
Thrombocytopaenia	WAS	707CtoG | A236G	rs132630270	X	48545317	48545317
Wiskott-Aldrich syndrome	WAS	714delT | 714del1 | 714del	N/A	X	48545324	48545324
Wiskott-Aldrich syndrome	WAS	723delC | 723del1 | 723del	N/A	X	48545333	48545333
Thrombocytopaenia	WAS	724AtoT | S242C	N/A	X	48545334	48545334
Wiskott-Aldrich syndrome	WAS	724delA | 724del1 | 724del	N/A	X	48545334	48545334
Wiskott-Aldrich syndrome	WAS	729delA | 729del1 | 729del	N/A	X	48545339	48545339
Wiskott-Aldrich syndrome	WAS	482dupC | 482dup1 | 482dup	N/A	X	48545344	48545345
Wiskott-Aldrich syndrome	WAS	734plus1GtoA	N/A	X	48545345	48545345
Wiskott-Aldrich syndrome	WAS	734plus1GtoT	N/A	X	48545345	48545345
Thrombocytopaenia	WAS	734plus5GtoA	N/A	X	48545349	48545349
Thrombocytopaenia	WAS	735minus25AtoC	N/A	X	48546418	48546418
Wiskott-Aldrich syndrome	WAS	735minus1GtoA	N/A	X	48546442	48546442
Wiskott-Aldrich syndrome	WAS	735delG | 735del1 | 735del	N/A	X	48546443	48546443
Wiskott-Aldrich syndrome	WAS	735_753delGCATGTCAGCCACGTGGGG | 735_753del19 | 735_753del	N/A	X	48546443	48546461
Wiskott-Aldrich syndrome	WAS	756GtoA | W252X	N/A	X	48546464	48546464
Wiskott-Aldrich syndrome	WAS	757delG | 757del1 | 757del	N/A	X	48546465	48546465
Wiskott-Aldrich syndrome	WAS	777plus1GtoA	N/A	X	48546486	48546486
Wiskott-Aldrich syndrome	WAS	777plus1GtoC	N/A	X	48546486	48546486
Wiskott-Aldrich syndrome	WAS	777+2_777+3delTG | 777+2_777+3del2 | 777+2_777+3del	N/A	X	48546487	48546488
Wiskott-Aldrich syndrome	WAS	777+2dupT | 777+2dup1 | 777+2dup	N/A	X	48546487	48546488
Wiskott-Aldrich syndrome	WAS	777+3_777+6delGAGT | 777+3_777+6del4 | 777+3_777+6del	N/A	X	48546488	48546491
Wiskott-Aldrich syndrome	WAS	777plus5GtoA	N/A	X	48546490	48546490
Wiskott-Aldrich syndrome	WAS	777plus5GtoC	N/A	X	48546490	48546490
Wiskott-Aldrich syndrome	WAS	1013_1014delGT | 1013_1014del2 | 1013_1014del	N/A	X	48547130	48547131
Wiskott-Aldrich syndrome	WAS	1016CtoA | S339Y	N/A	X	48547133	48547133
Wiskott-Aldrich syndrome	WAS	1019dupG | 1019dup1 | 1019dup	N/A	X	48547136	48547137
Wiskott-Aldrich syndrome	WAS	1020dupT | 1020dup1 | 1020dup	N/A	X	48547137	48547138
Wiskott-Aldrich syndrome	WAS	1022dupC | 1022dup1 | 1022dup	N/A	X	48547139	48547140
Wiskott-Aldrich syndrome	WAS	1031delC | 1031del1 | 1031del	N/A	X	48547148	48547148
Wiskott-Aldrich syndrome	WAS	1032delT | 1032del1 | 1032del	N/A	X	48547149	48547149
Wiskott-Aldrich syndrome	WAS	1037delC | 1037del1 | 1037del	N/A	X	48547154	48547154
Wiskott-Aldrich syndrome	WAS	1047dupT | 1047dup1 | 1047dup	N/A	X	48547164	48547165
Wiskott-Aldrich syndrome	WAS	1052delC | 1052del1 | 1052del	N/A	X	48547169	48547169
Wiskott-Aldrich syndrome	WAS	1058delC | 1058del1 | 1058del	N/A	X	48547175	48547175
Wiskott-Aldrich syndrome	WAS	1073_1074delGA | 1073_1074del2 | 1073_1074del	N/A	X	48547190	48547191
Wiskott-Aldrich syndrome	WAS	1073dupG | 1073dup1 | 1073dup	N/A	X	48547190	48547191
Wiskott-Aldrich syndrome	WAS	1075_1076delCCinsA | 1075_1076del2ins1 | 1075_1076delins	N/A	X	48547192	48547193
Wiskott-Aldrich syndrome	WAS	1079delC | 1079del1 | 1079del	N/A	X	48547196	48547196
Wiskott-Aldrich syndrome	WAS	1081CtoA | P361T	N/A	X	48547198	48547198
Thrombocytopaenia	WAS	1085delC | 1085del1 | 1085del	N/A	X	48547202	48547202
Thrombocytopaenia	WAS	1086delA | 1086del1 | 1086del	N/A	X	48547203	48547203
Wiskott-Aldrich syndrome	WAS	1090CtoT | R364X	N/A	X	48547207	48547207
Night blindness, congenital stationary, incomp	CACNA1F	2914CtoT | R972X	N/A	X	49074961	49074961
Night blindness, congenital stationary, incomp	CACNA1F	2905CtoT | R969X	rs122456134	X	49074970	49074970
Night blindness, congenital stationary, incomp	CACNA1F	584dupG | 584dup1 | 584dup	N/A	X	49087008	49087009
IPEX syndrome	FOXP3	304_305delTT | 304_305del2 | 304_305del	N/A	X	49114131	49114132
IPEX syndrome	FOXP3	227delT | 227del1 | 227del	N/A	X	49114209	49114209
Cornelia de Lange syndrome	SMC1A	3367CtoT | R1123W	N/A	X	53409223	53409223
Cornelia de Lange syndrome	SMC1A	3364TtoC | F1122L	N/A	X	53409226	53409226
Cornelia de Lange syndrome	SMC1A	1487GtoA | R496H	rs122454123	X	53436051	53436051
Cornelia de Lange syndrome	SMC1A	1486CtoT | R496C	N/A	X	53436052	53436052
Sideroblastic anaemia	ALAS2	1355GtoA | R452H	N/A	X	55041262	55041262
Sideroblastic anaemia	ALAS2	1354CtoA | R452S	N/A	X	55041263	55041263
Sideroblastic anaemia	ALAS2	1354CtoG | R452G	N/A	X	55041263	55041263
Sideroblastic anaemia	ALAS2	1354CtoT | R452C	rs137852311	X	55041263	55041263
Sideroblastic anaemia	ALAS2	1343GtoA | R448Q	N/A	X	55041274	55041274
Sideroblastic anaemia	ALAS2	1306CtoT | R436W	N/A	X	55041311	55041311
Sideroblastic anaemia	ALAS2	1276AtoG | M426V	N/A	X	55041341	55041341
Sideroblastic anaemia	ALAS2	1247GtoA | G416D	N/A	X	55041370	55041370
Sideroblastic anaemia	ALAS2	1232GtoA | R411H	N/A	X	55041385	55041385
Sideroblastic anaemia	ALAS2	1231CtoT | R411C	rs137852305	X	55041386	55041386
Choroideraemia	CHM	49plus5GtoT	N/A	X	85302483	85302483
Choroideraemia	CHM	17delC | 17del1 | 17del	N/A	X	85302520	85302520
Choroideraemia	CHM	3GtoT | M1I	N/A	X	85302534	85302534
Agammaglobulinaemia	BTK	1177+2delT | 1177+2del1 | 1177+2del	N/A	X	100612495	100612495
Agammaglobulinaemia	BTK	1176CtoA | Y392X	N/A	X	100612498	100612498
Agammaglobulinaemia	BTK	1173dupA | 1173dup1 | 1173dup	N/A	X	100612500	100612501
Agammaglobulinaemia	BTK	1173AtoG	N/A	X	100612501	100612501
Agammaglobulinaemia	BTK	1171GtoT | G391X	N/A	X	100612503	100612503
Agammaglobulinaemia	BTK	1166dupG | 1166dup1 | 1166dup	N/A	X	100612507	100612508
Agammaglobulinaemia	BTK	1157_1161delCCACT | 1157_1161del5 | 1157_1161del	N/A	X	100612513	100612517
Agammaglobulinaemia	BTK	1154delC | 1154del1 | 1154del	N/A	X	100612520	100612520
Agammaglobulinaemia	BTK	1145_1147delAGAinsTT | 1145_1147del3ins2 | 1145_1147delins	N/A	X	100612527	100612529
Agammaglobulinaemia	BTK	1138CtoT | Q380X	N/A	X	100612536	100612536
Alport syndrome	COL4A5	81plus4AtoG	N/A	X	107683440	107683440
Alport syndrome	COL4A5	1276GtoA | G426R	rs104886111	X	107834398	107834398
Alport syndrome	COL4A5	2782CtoT | Q928X	N/A	X	107865920	107865920
Alport syndrome	COL4A5	2788CtoT | Q930X	rs104886194	X	107865926	107865926
Alport syndrome	COL4A5	2802dupT | 2802dup1 | 2802dup	N/A	X	107865940	107865941
Alport syndrome	COL4A5	2804GtoA | G935D	rs104886195	X	107865942	107865942
Alport syndrome	COL4A5	2821GtoT | G941C	rs104886196	X	107865959	107865959
Alport syndrome	COL4A5	3016plus2TtoG	N/A	X	107867566	107867566
Alport syndrome	COL4A5	3535GtoA | G1179R	rs104886240	X	107909806	107909806
Alport syndrome	COL4A5	3538CtoT | Q1180X	rs104886241	X	107909809	107909809

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Alport syndrome	COL4A5	3543_3549delGGGTGAA | 3543_3549del7 | 3543_3549del	N/A	X	107909814	107909820
Alport syndrome	COL4A5	3544GtoC | G1182R	rs104886242	X	107909815	107909815
Simpson-Golabi-Behmel syndrome	GPC3	1159CtoT | R387X	rs122453121	X	132833930	132833930
Mental retardation, X-linked	SLC9A6	467TtoC | L156P	N/A	X	135080304	135080304
Reducing body myopathy	FHL1	310TtoC | C104R	rs122459147	X	135289328	135289328
Reducing body myopathy	FHL1	311GtoA | C104Y	N/A	X	135289329	135289329
Mucopolysaccharidosis II	IDS	213CtoA | S71R	N/A	X	148585714	148585714
Mucopolysaccharidosis II	IDS	213CtoG | S71R	N/A	X	148585714	148585714
Mucopolysaccharidosis II	IDS	212GtoA | S71N	rs113993954	X	148585715	148585715
Mucopolysaccharidosis II	IDS	208dupC | 208dup1 | 208dup	N/A	X	148585718	148585719
Mucopolysaccharidosis II	IDS	203CtoA | A68E	N/A	X	148585724	148585724
Mucopolysaccharidosis II	IDS	200TtoC | L67P	N/A	X	148585727	148585727
Mucopolysaccharidosis II	IDS	196CtoT | Q66X	N/A	X	148585731	148585731
Mental retardation	SLC6A8	11AtoG | K4R	N/A	X	152954040	152954040
Creatine deficiency	SLC6A8	1596plus1GtoA	N/A	X	152960089	152960089
Creatine deficiency	SLC6A8	1607_1612delTCTTCA | 1607_1612del6 | 1607_1612del	N/A	X	152960184	152960189
Barth syndrome	TAZ	710_711delTG | 710_711del2 | 710_711del	N/A	X	153649007	153649008
Cardiomyopathy, X-linked infantile	TAZ	718GtoA | G240R	N/A	X	153649015	153649015
Barth syndrome	TAZ	747_748insCGGGCGCCGGCGGCTCTCC | 747_748ins19 | 747_748ins	N/A	X	153649044	153649045
Dyskeratosis congenita with Hoyeraal-Hreidar	DKC1	941AtoG | K314R	N/A	X	153999059	153999059
Dyskeratosis congenita, X-linked	DKC1	949CtoG | L317V	N/A	X	153999067	153999067
Dyskeratosis congenita, X-linked	DKC1	949CtoT | L317F	rs121912290	X	153999067	153999067
Dyskeratosis congenita, X-linked	DKC1	961CtoG | L321V	rs2728726	X	153999079	153999079
Dyskeratosis congenita, X-linked	DKC1	965GtoA | R322Q	rs121912291	X	153999083	153999083
Haemophilia A	F8	5348GtoC | R1783T	N/A	X	154134720	154134720
Haemophilia A	F8	5345TtoG | I1782R	N/A	X	154134723	154134723
Haemophilia A	F8	5343TtoA | Y1781X	N/A	X	154134725	154134725
Haemophilia A	F8	5339CtoA | P1780Q	N/A	X	154134729	154134729
Haemophilia A	F8	5339CtoG | P1780R	N/A	X	154134729	154134729
Haemophilia A	F8	5339CtoT | P1780L	N/A	X	154134729	154134729
Haemophilia A	F8	1700TtoC | I567T	N/A	X	154185284	154185284
Haemophilia A	F8	1699AtoT | I567F	N/A	X	154185285	154185285
Haemophilia A	F8	1696CtoT | L566F	N/A	X	154185288	154185288
Haemophilia A	F8	1693GtoA | G565R	N/A	X	154185291	154185291
Haemophilia A	F8	1688CtoG | A563G	N/A	X	154185296	154185296
Haemophilia A	F8	1685TtoC | L562P	N/A	X	154185299	154185299
Haemophilia A	F8	1683TtoA | D561E	N/A	X	154185301	154185301
Haemophilia A	F8	1682AtoC | D561A	N/A	X	154185302	154185302
Haemophilia A	F8	1682AtoG | D561G	rs28937281	X	154185302	154185302
Haemophilia A	F8	1682delA | 1682del1 | 1682del	N/A	X	154185302	154185302
Haemophilia A	F8	1681GtoA | D561N	N/A	X	154185303	154185303
Haemophilia A	F8	1681GtoC | D561H	N/A	X	154185303	154185303
Haemophilia A	F8	1681GtoT | D561Y	N/A	X	154185303	154185303
Haemophilia A	F8	1679GtoC | R560T	N/A	X	154185305	154185305
Haemophilia A	F8	1678AtoG | R560G	N/A	X	154185306	154185306
Haemophilia A	F8	1675GtoT | E559X	N/A	X	154185309	154185309
Gonadal dysgenesis	SRY	488TtoA | L163X	N/A	Y	2655157	2655157
XY sex reversal	SRY	474dupA | 474dup1 | 474dup	N/A	Y	2655170	2655171
Gonadal dysgenesis	SRY	474delA | 474del1 | 474del	N/A	Y	2655171	2655171
Gonadal dysgenesis	SRY	427AtoG | S143G	N/A	Y	2655218	2655218
XY sex reversal	SRY	427AtoT | S143C	N/A	Y	2655218	2655218
Gonadal dysgenesis	SRY	407_415delAGATGCTGCinsGT | 407_415del9ins2 | 407_415delins	N/A	Y	2655230	2655238
XY sex reversal	SRY	397CtoG | R133G	N/A	Y	2655248	2655248
Gonadal dysgenesis	SRY	397CtoT | R133W	rs104894976	Y	2655248	2655248
Gonadal dysgenesis	SRY	394CtoG | R132G	N/A	Y	2655251	2655251
XY sex reversal	SRY	392CtoG | P131R	N/A	Y	2655253	2655253
XY sex reversal	SRY	388CtoT | R130X	N/A	Y	2655257	2655257
XY sex reversal	SRY	387TtoA | Y129X	N/A	Y	2655258	2655258
XY sex reversal	SRY	387TtoG | Y129X	N/A	Y	2655258	2655258
Gonadal dysgenesis	SRY	385TtoA | Y129N	N/A	Y	2655260	2655260
46,XY disorder of sex development with bilate	SRY	383AtoG | K128R	N/A	Y	2655262	2655262
Gonadal dysgenesis	SRY	381TtoA | Y127X	N/A	Y	2655264	2655264
XY sex reversal	SRY	380AtoG | Y127C	N/A	Y	2655265	2655265
XY sex reversal	SRY	380AtoT | Y127F	rs104894973	Y	2655265	2655265
XY sex reversal	SRY	379delT | 379del1 | 379del	N/A	Y	2655266	2655266
XY sex reversal	SRY	374CtoT | P125L	N/A	Y	2655271	2655271
Gonadal dysgenesis	SRY	364_367delGAGA | 364_367del4 | 364_367del	N/A	Y	2655278	2655281
XY sex reversal	SRY	352GtoC | A118P	N/A	Y	2655293	2655293
Gonadal dysgenesis	SRY	347TtoC | L116S	N/A	Y	2655298	2655298
Gonadal dysgenesis	SRY	337GtoA | A113T	rs104894966	Y	2655308	2655308
Gonadal dysgenesis	SRY	334GtoA | E112K	N/A	Y	2655311	2655311
Gonadal dysgenesis	SRY	326TtoC | F109S	rs104894956	Y	2655319	2655319
Gonadal dysgenesis	SRY	324delA | 324del1 | 324del	N/A	Y	2655321	2655321
Gonadal dysgenesis	SRY	323CtoG | P108R	N/A	Y	2655322	2655322
XY sex reversal	SRY	320GtoA | W107X	rs104894967	Y	2655325	2655325
Primary amenorrhea	SRY	318dupA | 318dup1 | 318dup	N/A	Y	2655326	2655327
Gonadal dysgenesis	SRY	317AtoT | K106I	rs104894964	Y	2655328	2655328
Hermaphroditism	SRY	302TtoA | L101H	N/A	Y	2655343	2655343
XY sex reversal	SRY	301CtoG | L101V	N/A	Y	2655344	2655344
Gonadal dysgenesis	SRY	294GtoA | W98X	N/A	Y	2655351	2655351
Primary amenorrhea	SRY	292TtoC | W98R	N/A	Y	2655353	2655353
XY sex reversal	SRY	289CtoT | Q97X	N/A	Y	2655356	2655356
Gonadal dysgenesis	SRY	287AtoG | Y96C	N/A	Y	2655358	2655358
Gonadal dysgenesis	SRY	284GtoA | G95E	rs104894972	Y	2655361	2655361
Gonadal dysgenesis	SRY	283GtoC | G95R	rs104894974	Y	2655362	2655362
Gonadal dysgenesis	SRY	281TtoC | L94P	N/A	Y	2655364	2655364
Noonan syndrome	SRY	280delC | 280del1 | 280del	N/A	Y	2655365	2655365
XY sex reversal	SRY	277CtoT | Q93X	rs104894958	Y	2655368	2655368
XY sex reversal	SRY	275AtoT | K92M	N/A	Y	2655370	2655370
XY sex reversal	SRY	274AtoT | K92X	rs104894970	Y	2655371	2655371
XY sex reversal	SRY	271AtoG | S91G	N/A	Y	2655374	2655374
Gonadal dysgenesis	SRY	270CtoG | I90M	rs104894959	Y	2655375	2655375
46,XY disorder of sex development	SRY	265GtoA | E89K	N/A	Y	2655380	2655380
XY sex reversal	SRY	264dupA | 264dup1 | 264dup	N/A	Y	2655380	2655381
XY sex reversal	SRY	259AtoT | N87Y	N/A	Y	2655386	2655386

disease	gene	hgvsAll	dbsnp	chromosome	startCoord	endCoord
Gonadal dysgenesis	SRY	256CtoT | R86X	N/A	Y	2655389	2655389
XY sex reversal	SRY	254TtoC | M85T	N/A	Y	2655391	2655391
Gonadal dysgenesis	SRY	245delA | 245del1 | 245del	N/A	Y	2655400	2655400
XY sex reversal	SRY	243_244insT | 243_244ins1 | 243_244ins	N/A	Y	2655401	2655402
Hermaphroditism	SRY	237TtoC | A79A	N/A	Y	2655408	2655408
Gonadal dysgenesis	SRY	233TtoC | M78T	N/A	Y	2655412	2655412
Gonadal dysgenesis	SRY	228dupC | 228dup1 | 228dup	N/A	Y	2655416	2655417
XY sex reversal	SRY	222GtoC | Q74H	N/A	Y	2655423	2655423
Gonadal dysgenesis	SRY	220CtoT | Q74X	N/A	Y	2655425	2655425
Gonadal dysgenesis	SRY	214CtoG | R72G	N/A	Y	2655431	2655431
Gonadal dysgenesis	SRY	209GtoA | W70X	rs104894965	Y	2655436	2655436
Gonadoblastoma	SRY	209GtoT | W70L	N/A	Y	2655436	2655436
Gonadal dysgenesis	SRY	203TtoC | I68T	rs104894968	Y	2655442	2655442
Gonadal dysgenesis	SRY	199TtoC | F67L	N/A	Y	2655446	2655446
Gonadal dysgenesis	SRY	199TtoG | F67V	N/A	Y	2655446	2655446
Gonadal dysgenesis	SRY	193AtoC | N65H	N/A	Y	2655452	2655452
Gonadal dysgenesis	SRY	192GtoA | M64I	rs104894969	Y	2655453	2655453
Gonadal dysgenesis	SRY	191TtoG | M64R	N/A	Y	2655454	2655454
Gonadal dysgenesis	SRY	184CtoG | R62G	N/A	Y	2655461	2655461
Hermaphroditism	SRY	179TtoC | V60A	N/A	Y	2655466	2655466
Gonadal dysgenesis	SRY	178GtoC | V60L	rs104894957	Y	2655467	2655467
Gonadal dysgenesis	SRY	175AtoG | R59G	N/A	Y	2655470	2655470
XY sex reversal	SRY	170AtoG | Q57R	N/A	Y	2655475	2655475
Gonadal dysgenesis	SRY	126dupT | 126dup1 | 126dup	N/A	Y	2655518	2655519
XY sex reversal	SRY	113AtoG | E38G	N/A	Y	2655532	2655532
Gonadal dysgenesis	SRY	89GtoT | R30I	N/A	Y	2655556	2655556
Testicular dysgenesis syndrome	SRY	71delA | 71del1 | 71del	N/A	Y	2655574	2655574
XY sex reversal	SRY	53GtoA | S18N	rs104894971	Y	2655592	2655592
XY sex reversal	SRY	38dupA | 38dup1 | 38dup	N/A	Y	2655606	2655607
Gonadal dysgenesis	SRY	12TtoA | Y4X	rs104894975	Y	2655633	2655633
Gonadal dysgenesis	SRY	12delT | 12del1 | 12del	N/A	Y	2655633	2655633
Gonadal dysgenesis	SRY	8CtoT | S3L	N/A	Y	2655637	2655637
XY sex reversal	SRY	4CtoT | Q2X	rs104894977	Y	2655641	2655641
Gonadal dysgenesis	SRY	NULL	N/A	Y	2655719	2655719
XY sex reversal	SRY	NULL	N/A	Y	2655774	2655774
Coarctation of the aorta, non-syndromic	TBL1Y	205GtoC | D69H	N/A	Y	6931938	6931938
Azoospermia	USP9Y	773+3_773+6delAAGT | 773+3_773+6del4 | 773+3_773+6del	N/A	Y	14847664	14847667