Amby Lab

Carrier status testing SNPs

Amby Lab is making high-quality genetic Carrier Status testing applying SNP microarray genotyping to identify alterations in human genome.

Amby Lab identifies alterations in the SNPs listed below.
This list contains SNPs, for which all detectable mutations for every disease are responsible for more than 50% of the cases.

SNP LIST							
DISEASE	**CHR.**	**GENOMIC COORD.**	**GENE**	**EXON**	**MUTATION**	**DBSNP ID**	**OMIM ID**
Abetalipoproteinemia	4	Chr4:100543913	MTTP	18	c.2674G>T	rs146064714	200100
Aceruloplasminemia	3	Chr3:148897374	CP	15	c.2630G>A	rs121909579	604290
Achondrogenesis, type ib	5	Chr5:149357747	SLC26A2	2	c.532C>T	rs104893919	600972
Achromatopsia 3	8	Chr8:87656008	CNGB3	10	c.1148delC	-	262300
Achromatopsia 3	8	Chr8:87680283	CNGB3	5	c.607C>T	rs267606739	262300
Acth deficiency, isolated	1	Chr1:168274374	TBX19	6	c.856C>T	rs74315376	201400
Acyl-coa dehydrogenase, short-chain, deficiency of	12	Chr12:121174897	ACADS	3	c.319C>T	rs61732144	201470
Acyl-coa dehydrogenase, short-chain, deficiency of	12	Chr12:121176971	ACADS	9	c.1058C>T	rs28941773	201470
Albinism, oculocutaneous, type 1	11	Chr11:88911261	TYR	1	c.140G>A	rs61753180	203100
Albinism, oculocutaneous, type ii	15	Chr15:28171315	OCA2	19	c.2037G>C	rs121918169	203200
Albinism, oculocutaneous, type iii	9	Chr9:12695626	TYRP1	3	c.497C>G	rs104894130	203290
Albinism, oculocutaneous, type iii	9	Chr9:12704544	TYRP1	6	c.1103delA	-	203290
Alkaptonuria	3	Chr3:120363252	HGD	10	c.688C>T	rs28942100	203500
Alkaptonuria	3	Chr3:120393748	HGD	3	c.175delA	-	203500
Alkaptonuria	3	Chr3:120393784	HGD	3	c.140C>A	rs369517993	203500
Alpha-thalassemia	11	Chr11:5248004	HBB	2	c.118C>T	rs76728603	604131
Alpha-thalassemia	X	ChrX:76776310	ATRX	34	c.7156C>T	rs122445099	301040
Amegakaryocytic thrombocytopenia, congenital	1	Chr1:43804305	MPL	3	c.305G>C	rs28928907	604498
Amelogenesis imperfecta, hypomaturation type, iia1	19	Chr19:51411852	KLK4	3	c.458G>A	rs104894704	204700
Amelogenesis imperfecta, type ig	17	Chr17:66551883	FAM20A	2	c.406C>T	rs144411158	614253
Amyloidosis, familial visceral	4	Chr4:155506863	FGA	5	c.1718G>A	rs78506343	105200
Amyotrophic lateral sclerosis 9	14	Chr14:21161845	ANG	2	c.122A>T	rs121909536	611895
Argininemia	6	Chr6:131902418	ARG1	4	c.389G>A	rs104893947	207800
Argininemia	6	Chr6:131904950	ARG1	8	c.895C>T	rs104893940	207800
Arterial calcification, generalized, of infancy, 2	16	Chr16:16256935	ABCC6	24	c.3421C>T	rs72653706	614473
Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	9	Chr9:32974493	APTX	7	c.879G>A	rs104894103	208920
Bardet-biedl syndrome 1	11	Chr11:66299163	BBS1	16	c.1645G>T	rs121917777	209900
Bardet-biedl syndrome 1	20	Chr20:10393286	MKKS	3	c.873_876dup	rs113994196	209900

Bardet-biedl syndrome 1	20	Chr20:10393721	MKKS	3	c.431_441delTr	s113994195	209900
Bardet-biedl syndrome 1	4	Chr4:123663370	BBS12	3	c.323C>G	rs151344630	209900
Bardet-biedl syndrome 2	16	Chr16:56536702	BBS2	8	c.823C>T	rs121908177	615981
Bardet-biedl syndrome 2	16	Chr16:56548535	BBS2	2	c.175C>T	rs121908176	615981
Bardet-biedl syndrome 3	3	Chr3:97506848	ARL6	6	c.364C>T	rs104893678	600151
Bietti crystalline corneoretinal dystrophy	4	Chr4:187117229	CYP4V2	3	c.400G>T	rs199476189	210370
Bietti crystalline corneoretinal dystrophy	4	Chr4:187126386	CYP4V2	8	c.1020G>A	rs199476198	210370
Bietti crystalline corneoretinal dystrophy	4	Chr4:187130126	CYP4V2	9	c.1198C>T	rs138444697	210370
Bietti crystalline corneoretinal dystrophy	4	Chr4:187130127	CYP4V2	9	c.1199G>A	rs199476203	210370
Bile acid synthesis defect, congenital, 3	8	Chr8:65517310	CYP7B1	5	c.1162C>T	rs72554620	613812
Blood group, junior system	4	Chr4:89039366	ABCG2	7	c.736C>T	rs200190472	614490
Blood group, junior system	4	Chr4:89039396	ABCG2	7	c.706C>T	rs140207606	614490
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32893420	BRCA2	3	c.274C>T	rs80358529	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32900690	BRCA2	7	c.574_575delA	rs80359533	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32900701	BRCA2	7	c.582G>A	rs80358810	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32903604	BRCA2	8	c.658_659delGr	s80359604	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32905062	BRCA2	9	c.688A>T	rs80358913	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32905069	BRCA2	9	c.700delT	rs80359633	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32905148	BRCA2	9	c.778_779delGr	s80359680	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32906852	BRCA2	10	c.1238delT	rs80359271	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32906871	BRCA2	10	c.1257delT	rs80359272	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32906907	BRCA2	10	c.1296_1297d	rs80359276	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32907365	BRCA2	10	c.1755_1759d	rs80359302	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32907447	BRCA2	10	c.1832C>A	rs80358474	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32910420	BRCA2	11	c.1929delG	rs80359316	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32910583	BRCA2	11	c.2092delC	rs80359322	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32910901	BRCA2	11	c.2409T>G	rs80358504	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32911297	BRCA2	11	c.2808_2811d	rs80359352	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32911310	BRCA2	11	c.2818C>T	rs80358532	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32911471	BRCA2	11	c.2979G>A	rs80358544	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32911601	BRCA2	11	c.3109C>T	rs80358557	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32911650	BRCA2	11	c.3158T>G	rs41293477	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32911658	BRCA2	11	c.3170_3174d	rs80359373	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912035	BRCA2	11	c.3545_3546d	rs80359388	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912089	BRCA2	11	c.3599_3600d	rs80359391	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912180	BRCA2	11	c.3689delC	rs80359398	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912206	BRCA2	11	c.3717delA	rs80359401	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912337	BRCA2	11	c.3847_3848d	rs80359405	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912414	BRCA2	11	c.3922G>T	rs80358638	612555

Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912714	BRCA2	11	c.4222C>T	rs80358663	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912767	BRCA2	11	c.4276dupA	rs80359438	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912770	BRCA2	11	c.4284dupT	rs80359440	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912940	BRCA2	11	c.4449delA	rs80359448	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32912963	BRCA2	11	c.4472_4475d	rs80359452	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32913125	BRCA2	11	c.4638delT	rs80359462	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32913140	BRCA2	11	c.4648G>T	rs80358695	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32913365	BRCA2	11	c.4876_4877d	rs80359470	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32913702	BRCA2	11	c.5213_5216d	rs80359493	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32913757	BRCA2	11	c.5266_5269d	rs80359501	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32913777	BRCA2	11	c.5290_5291d	rs80359503	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32913794	BRCA2	11	c.5303_5304d	rs80359505	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32913970	BRCA2	11	c.5482_5486d	rs80359516	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914065	BRCA2	11	c.5576_5579d	rs80359521	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914106	BRCA2	11	c.5614A>T	rs80358783	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914109	BRCA2	11	c.5621_5624d	rs80359526	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914137	BRCA2	11	c.5645C>A	rs80358785	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914288	BRCA2	11	c.5799_5802d	rs80359538	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914319	BRCA2	11	c.5828delC	rs80359541	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914437	BRCA2	11	c.5946delT	rs80359549	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914472	BRCA2	11	c.5980C>T	rs80358831	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914698	BRCA2	11	c.6206T>G	rs80358859	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914859	BRCA2	11	c.6373dupA	rs80359577	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914935	BRCA2	11	c.6444dupT	rs80359590	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914953	BRCA2	11	c.6468_6469d	rs80359597	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32914973	BRCA2	11	c.6486_6489d	rs80359598	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32920966	BRCA2	13	c.6941delC	rs80359628	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32920967	BRCA2	13	c.6944_6947d	rs80359629	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32920978	BRCA2	13	c.6952C>T	rs80358920	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32929123	BRCA2	14	c.7133C>G	rs276174889	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32929366	BRCA2	14	c.7379_7382d	rs80359648	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32929400	BRCA2	14	c.7412_7421d	rs80359649	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32930609	BRCA2	15	c.7480C>T	rs80358972	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32930687	BRCA2	15	c.7558C>T	rs80358981	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32930690	BRCA2	15	c.7567_7568d	rs80359664	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32936711	BRCA2	17	c.7857G>A	rs80359011	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32936732	BRCA2	17	c.7878G>C	rs80359013	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32936828	BRCA2	17	c.7974C>G	rs80359025	612555

Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32937558	BRCA2	18	c.8219T>A	rs80359070	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32937635	BRCA2	18	c.8297delC	rs80359705	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32937659	BRCA2	18	c.8322dupT	rs80359706	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32945105	BRCA2	20	c.8501delC	rs80359712	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32945179	BRCA2	20	c.8575delC	rs80359718	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32945189	BRCA2	20	c.8585dupT	rs80359720	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32953568	BRCA2	22	c.8869C>T	rs276174913	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32953650	BRCA2	22	c.8951C>G	rs80359146	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32953957	BRCA2	23	c.9026_9030d	rs80359741	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32954180	BRCA2	24	c.9154C>T	rs45580035	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32969000	BRCA2	25	c.9435_9436d	rs80359763	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32971132	BRCA2	26	c.9599C>G	rs80359230	612555
Breast-ovarian cancer, familial, susceptibility to, 2	13	Chr13:32972626	BRCA2	27	c.9976A>T	rs11571833	612555
C1q deficiency	1	Chr1:22965784	C1QA	3	c.622C>T	rs121909581	613652
Calcification of joints and arteries	6	Chr6:86181054	NT5E	3	c.662C>A	-	211800
Carbamoyl phosphate synthetase i deficiency, hyperammonemia due to	2	Chr2:211504769	CPS1	25	c.2963G>T	rs121912595	237300
Carnitine deficiency, systemic primary	5	Chr5:131714072	SLC22A5	2	c.396G>A	rs72552727	212140
Carnitine deficiency, systemic primary	5	Chr5:131719847	SLC22A5	3	c.506G>A	rs121908889	212140
Carnitine deficiency, systemic primary	5	Chr5:131722736	SLC22A5	5	c.844C>T	rs121908886	212140
Carnitine deficiency, systemic primary	5	Chr5:131729380	SLC22A5	9	c.1463G>A	rs28383481	212140
Carpenter syndrome 1	6	Chr6:57059615	RAB23	5	c.434T>A	rs121908171	201000
Ceroid lipofuscinosis, neuronal, 1	1	Chr1:40546155	PPT1	6	c.541G>A	rs148412181	256730
Ceroid lipofuscinosis, neuronal, 1	1	Chr1:40555128	PPT1	5	c.490C>T	-	256730
Ceroid lipofuscinosis, neuronal, 1	1	Chr1:40555167	PPT1	5	c.451C>T	rs137852700	256730
Ceroid lipofuscinosis, neuronal, 1	1	Chr1:40562882	PPT1	1	c.29T>A	rs137852699	256730
Ceroid lipofuscinosis, neuronal, 2	11	Chr11:6638271	TPP1	6	c.622C>T	rs119455955	204500
Charcot-marie-tooth disease, 4c	5	Chr5:148389835	SH3TC2	14	c.3325C>T	rs80338934	601596
Charcot-marie-tooth disease, 4c	5	Chr5:148406466	SH3TC2	11	c.2829T>G	rs80338932	601596
Charcot-marie-tooth disease, 4c	5	Chr5:148406585	SH3TC2	11	c.2710C>T	rs80338931	601596
Charcot-marie-tooth disease, 4c	X	ChrX:70444347	GJB1	2	c.790C>T	-	302800
Charcot-marie-tooth disease, axonal, type 2a2	1	Chr1:12069698	MFN2	17	c.2119C>T	rs119103267	609260
Charcot-marie-tooth disease, recessive intermediate c	1	Chr1:6528438	PLEKHG5	21	c.2695G>A	rs202191898	615376
Charcot-marie-tooth disease, x-linked dominant, 1	5	Chr5:148406435	SH3TC2	11	c.2860C>T	rs80338933	601596
Cholestasis, benign recurrent intrahepatic, 2	2	Chr2:169847329	ABCB11	9	c.890A>G	rs11568372	605479
Cholestasis, intrahepatic, of pregnancy 3	7	Chr7:87041264	ABCB4	23	c.2869C>T	rs121918440	614972
Cholestasis, intrahepatic, of pregnancy 3	7	Chr7:87082366	ABCB4	6	c.430C>T	rs72552780	614972
Cholestasis, progressive familial intrahepatic, 2	2	Chr2:169788931	ABCB11	24	c.3169C>T	rs72549397	601847
Cholestasis, progressive familial intrahepatic, 2	2	Chr2:169826641	ABCB11	15	c.1723C>T	rs72549401	601847

Cholestasis, progressive familial intrahepatic, 2	2	Chr2:169847329	ABCB11	9	c.890A>G	rs11568372	601847
Ciliary dyskinesia, primary, 19	8	Chr8:133645039	LRRC6	5	c.598_599delA	-	614935
Ciliary dyskinesia, primary, 19	8	Chr8:133645077	LRRC6	5	c.562C>T	-	614935
Citrullinemia, type ii, adult-onset	7	Chr7:95820501	SLC25A13	7	c.674C>A	rs80338719	603471
Citrullinemia, type ii, neonatal-onset	7	Chr7:95750995	SLC25A13	17	c.1816C>T	rs80338729	605814
Citrullinemia, type ii, neonatal-onset	7	Chr7:95813688	SLC25A13	11	c.1081C>T	rs80338721	605814
Citrullinemia, type ii, neonatal-onset	7	Chr7:95822414	SLC25A13	6	c.550C>T	rs80338716	605814
Coach syndrome	16	Chr16:53679807	RPGRIP1L	17	c.2413C>T	rs145665129	216360
Cockayne syndrome b	10	Chr10:50686483	ERCC6	11	c.2203C>T	rs121917901	133540
Combined oxidative phosphorylation deficiency 1	3	Chr3:158363475	GFM1	2	c.139C>T	rs119470019	609060
Combined oxidative phosphorylation deficiency 15	15	Chr15:65295576	MTFMT	9	c.994C>T	rs200286768	614947
Complement component 5 deficiency	9	Chr9:123725027	C5	36	c.4426C>T	rs121909588	609536
Complement component 5 deficiency	9	Chr9:123812470	C5	1	c.55C>T	rs121909587	609536
Complement component 9 deficiency	5	Chr5:39306855	C9	9	c.1280C>G	rs121909594	613825
Complement component 9 deficiency	5	Chr5:39341378	C9	4	c.346C>T	rs121909592	613825
Cone-rod dystrophy 3	1	Chr1:94473807	ABCA4	42	c.5882G>A	rs1800553	604116
Cone-rod dystrophy 3	1	Chr1:94544895	ABCA4	9	c.1222C>T	-	604116
Cystinosis, nephropathic	17	Chr17:3558349	CTNS	6	c.283G>T	rs121908124	219800
Cystinosis, nephropathic	17	Chr17:3558599	CTNS	7	c.414G>A	rs113994205	219800
Cystinuria	19	Chr19:33334838	SLC7A9	10	c.997C>T	rs121908484	220100
Cystinuria	19	Chr19:33355167	SLC7A9	4	c.313G>A	rs121908480	220100
Cystinuria	2	Chr2:44539792	SLC3A1	8	c.1400T>C	rs121912691	220100
De sanctis-cacchione syndrome	10	Chr10:50686483	ERCC6	11	c.2203C>T	rs121917901	278800
Deafness, autosomal dominant 6	4	Chr4:6303576	WFS1	8	c.2054G>A	rs142668478	600965
Deafness, autosomal recessive 12	10	Chr10:73553289	CDH23	46	c.6604G>A	rs121908349	601386
Deafness, autosomal recessive 18a	11	Chr11:17526211	USH1C	20	c.2167C>T	rs146451547	602092
Deafness, autosomal recessive 2	11	Chr11:76868046	MYO7A	7	c.731G>A	rs121965081	600060
Deafness, autosomal recessive 31	9	Chr9:117166262	DFNB31	10	c.2332C>T	rs137852839	607084
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	1	Chr1:160011281	KCNJ10	2	c.1042C>T	rs137853074	600791
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	7	Chr7:107302088	SLC26A4	2	c.2T>C	rs111033302	600791
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	7	Chr7:107312690	SLC26A4	4	c.412G>T	rs111033199	600791
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	7	Chr7:107315415	SLC26A4	6	c.626G>T	rs111033303	600791
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	7	Chr7:107329499	SLC26A4	9	c.1003T>C	rs111033212	600791
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	7	Chr7:107336429	SLC26A4	13	c.1489G>A	rs111033308	600791
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	7	Chr7:107336480	SLC26A4	13	c.1540C>A	rs121908366	600791
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	7	Chr7:107338530	SLC26A4	14	c.1588T>C	rs111033254	600791

Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	7	Chr7:107342483	SLC26A4	17	c.2015G>A	rs111033309	600791
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	7	Chr7:107350577	SLC26A4	19	c.2168A>G	rs121908362	600791
Deafness, autosomal recessive 59	2	Chr2:179320828	DFNB59	4	c.499C>T	rs118203989	610220
Deafness, autosomal recessive 7	9	Chr9:75309494	TMC1	7	c.100C>T	rs121908073	600974
Deafness, autosomal recessive 7	9	Chr9:75404174	TMC1	15	c.1165C>T	rs151001642	600974
Deafness, autosomal recessive 7	9	Chr9:75406910	TMC1	16	c.1333C>T	rs372710475	600974
Deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures syndrome	16	Chr16:2546207	TBC1D24	2	c.58C>T	rs201257588	222760
Diastrophic dysplasia	5	Chr5:149357747	SLC26A2	2	c.532C>T	rs104893919	222600
Dubin-johnson syndrome	10	Chr10:101591826	ABCC2	23	c.3196C>T	rs72558199	237500
Ectopia lentis 2, isolated, autosomal recessive	1	Chr1:150529450	ADAMTSL 4	12	c.1854T>G	rs118203985	225100
Enterokinase deficiency	21	Chr21:19685292	TMPRSS1 5	18	c.2135C>G	rs77200626	226200
Epidermolysis bullosa, junctional, herlitz type	1	Chr1:209791900	LAMB3	19	c.2806C>T	rs121912485	226700
Epidermolysis bullosa, junctional, herlitz type	1	Chr1:209799066	LAMB3	14	c.1903C>T	rs80356682	226700
Epidermolysis bullosa, junctional, herlitz type	1	Chr1:209799139	LAMB3	14	c.1830G>A	rs121912484	226700
Epidermolysis bullosa, junctional, herlitz type	1	Chr1:209806023	LAMB3	8	c.727C>T	rs80356681	226700
Epidermolysis bullosa, junctional, herlitz type	1	Chr1:209807860	LAMB3	6	c.496C>T	rs121912483	226700
Epidermolysis bullosa, junctional, herlitz type	1	Chr1:209823368	LAMB3	3	c.124C>T	rs80356680	226700
Epidermolysis bullosa, junctional, herlitz type	18	Chr18:21453158	LAMA3	1	c.151dupG	rs80356678	226700
Epidermolysis bullosa, junctional, herlitz type	18	Chr18:21487603	LAMA3	53	c.6808C>T	rs137852757	226700
Epidermolysis bullosa, junctional, herlitz type	18	Chr18:21487827	LAMA3	54	c.6943A>T	rs80356679	226700
Epidermolysis bullosa, junctional, non-herlitz type	1	Chr1:209799066	LAMB3	14	c.1903C>T	rs80356682	226650
Epidermolysis bullosa, junctional, non-herlitz type	10	Chr10:105794469	COL17A1	51	c.3676C>T	rs121912769	226650
Epidermolysis bullosa, junctional, non-herlitz type	18	Chr18:21519286	LAMA3	68	c.8962C>T	rs137852758	226650
Epilepsy, pyridoxine-dependent	5	Chr5:125887751	ALDH7A1	14	c.1279G>C	rs121912707	266100
Fabry disease	X	ChrX:100656740	GLA	3	c.427G>A	rs104894845	301500
Fabry disease	X	ChrX:100658833	GLA	2	c.335G>A	rs372966991	301500
Factor v deficiency	1	Chr1:169487691	F5	23	c.6304C>T	rs118203910	227400
Factor v deficiency	1	Chr1:169511927	F5	13	c.2401C>T	rs118203908	227400
Factor xi deficiency	4	Chr4:187195347	F11	5	c.403G>T	rs121965063	612416
Factor xi deficiency	4	Chr4:187201487	F11	9	c.976C>T	rs28934608	612416
Factor xiii, a subunit, deficiency of	6	Chr6:6152107	F13A1	14	c.1984C>T	rs267606789	613225
Familial cold autoinflammatory syndrome 2	19	Chr19:54314063	NLRP12	3	c.850C>T	rs104895564	611762
Fanconi anemia, complementation group c	9	Chr9:97864024	FANCC	15	c.1642C>T	rs104886457	227645
Fanconi anemia, complementation group c	9	Chr9:97912338	FANCC	7	c.553C>T	rs121917783	227645
Fanconi anemia, complementation group c	9	Chr9:98011506	FANCC	2	c.67delG	rs104886459	227645
Fanconi anemia, complementation group c	9	Chr9:98011537	FANCC	2	c.37C>T	rs121917784	227645
Fanconi anemia, complementation group i	15	Chr15:89858549	FANCI	37	c.3853C>T	rs121918164	609053
Fanconi anemia, complementation group i	15	Chr15:89858550	FANCI	37	c.3854G>A	rs121918163	609053

Fanconi anemia, complementation group j	17	Chr17:59793412	BRIP1	17	c.2392C>T	rs137852986	609054
Fructose intolerance, hereditary	9	Chr9:104187814	ALDOB	7	c.720C>A	rs118204426	229600
Fructose intolerance, hereditary	9	Chr9:104190766	ALDOB	4	c.360_363delC	-	229600
Fructose intolerance, hereditary	9	Chr9:104192183	ALDOB	3	c.178C>T	rs118204429	229600
Fructose intolerance, hereditary	9	Chr9:104193160	ALDOB	2	c.10C>T	rs118204428	229600
Galactokinase deficiency	17	Chr17:73754172	GALK1	8	c.1144C>T	rs111033608	230200
Gitelman syndrome	16	Chr16:56904031	SLC12A3	5	c.625C>T	rs28936388	263800
Gitelman syndrome	16	Chr16:56906649	SLC12A3	8	c.1046C>T	rs121909383	263800
Gitelman syndrome	16	Chr16:56918054	SLC12A3	14	c.1763C>T	rs121909382	263800
Gitelman syndrome	16	Chr16:56928506	SLC12A3	22	c.2612G>A	rs267607051	263800
Glanzmann thrombasthenia	17	Chr17:45384950	ITGB3	14	c.2248C>T	rs121918450	273800
Glaucoma 1, open angle, a	1	Chr1:171605478	MYOC	3	c.1102C>T	rs74315329	137750
Glaucoma 1, open angle, o	19	Chr19:49564639	NTF4	2	c.616C>T	rs121918427	613100
Glaucoma 3, primary congenital, a	2	Chr2:38302361	CYP1B1	2	c.171G>A	rs72549387	231300
Glaucoma 3, primary congenital, d	14	Chr14:75022332	LTBP2	4	c.895C>T	rs121918355	613086
Glutathione synthetase deficiency	20	Chr20:33530409	GSS	5	c.373C>T	rs28936396	266130
Glycogen storage disease 0, liver	12	Chr12:21721886	GYS2	5	c.736C>T	rs121918419	240600
Glycogen storage disease 1	11	Chr11:118895961	SLC37A4	11	c.1129G>T	rs121908975	232220
Glycogen storage disease 1	11	Chr11:118895980	SLC37A4	11	c.1108_1109d	rs80356491	232220
Glycogen storage disease 1	17	Chr17:41055964	G6PC	2	c.247C>T	rs1801175	232220
Glycogen storage disease 1	17	Chr17:41055965	G6PC	2	c.248G>A	rs1801176	232220
Glycogen storage disease 1	17	Chr17:41059575	G6PC	3	c.379_380dup	rs80356488	232220
Glycogen storage disease 1	17	Chr17:41061435	G6PC	4	c.562G>C	rs80356482	232220
Glycogen storage disease 1	17	Chr17:41063093	G6PC	5	c.724C>T	rs80356485	232220
Glycogen storage disease 1	17	Chr17:41063178	G6PC	5	c.809G>T	rs80356483	232220
Glycogen storage disease 1	17	Chr17:41063408	G6PC	5	c.1039C>T	rs80356487	232220
Glycogen storage disease ii	17	Chr17:78086721	GAA	14	c.1935C>A	rs28940868	232300
Glycogen storage disease ii	17	Chr17:78092070	GAA	18	c.2560C>T	rs121907943	232300
Glycogen storage disease ixb	16	Chr16:47630336	PHKB	13	c.1257T>A	rs121918021	261750
Glycogen storage disease ixb	16	Chr16:47730322	PHKB	29	c.2926G>T	rs199948078	261750
Glycogen storage disease v	11	Chr11:64517969	PYGM	17	c.2056G>A	rs144081869	232600
Glycogen storage disease v	11	Chr11:64525298	PYGM	5	c.613G>A	rs119103251	232600
Glycogen storage disease v	11	Chr11:64527223	PYGM	1	c.148C>T	rs116987552	232600
Glycogen storage disease x	7	Chr7:44104896	PGAM2	1	c.233G>A	rs10250779	261670
Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type ii	1	Chr1:183546802	NCF2	3	c.298C>T	rs119103276	233710
Hartnup disorder	5	Chr5:1213632	SLC6A19	5	c.718C>T	rs121434347	234500
Hemophagocytic lymphohistiocytosis, familial, 3	17	Chr17:73836398	UNC13D	10	c.766C>T	rs121434352	608898
Hemophilia b	X	ChrX:138619249	F9	2	c.169C>T	rs137852223	306900
Hemophilia b	X	ChrX:138619303	F9	2	c.223C>T	rs137852227	306900
Hemophilia b	X	ChrX:138619304	F9	2	c.224G>A	rs137852228	306900
Hemophilia b	X	ChrX:138619317	F9	2	c.237A>C	rs137852229	306900
Hemophilia b	X	ChrX:138623244	F9	4	c.287A>C	rs137852231	306900
Hemophilia b	X	ChrX:138623258	F9	4	c.301C>G	rs137852232	306900
Hemophilia b	X	ChrX:138623273	F9	4	c.316G>A	rs137852233	306900

Hemophilia b	X	ChrX:138623285	F9	4	c.328G>A	rs137852274	306900
Hemophilia b	X	ChrX:138630609	F9	5	c.479G>C	rs137852235	306900
Hemophilia b	X	ChrX:138630614	F9	5	c.484C>T	rs137852272	306900
Hemophilia b	X	ChrX:138633271	F9	6	c.571C>T	rs137852237	306900
Hemophilia b	X	ChrX:138633272	F9	6	c.572G>A	rs137852238	306900
Hemophilia b	X	ChrX:138633355	F9	6	c.655C>T	rs137852239	306900
Hemophilia b	X	ChrX:138633409	F9	6	c.709C>T	rs137852244	306900
Hemophilia b	X	ChrX:138642931	F9	7	c.755G>C	rs267606792	306900
Hemophilia b	X	ChrX:138642980	F9	7	c.804T>G	rs137852246	306900
Hemophilia b	X	ChrX:138643716	F9	8	c.872A>T	rs137852279	306900
Hemophilia b	X	ChrX:138643724	F9	8	c.880C>T	rs137852248	306900
Hemophilia b	X	ChrX:138643725	F9	8	c.881G>A	rs137852249	306900
Hemophilia b	X	ChrX:138643736	F9	8	c.892C>T	rs137852250	306900
Hemophilia b	X	ChrX:138643842	F9	8	c.998C>T	rs137852252	306900
Hemophilia b	X	ChrX:138643853	F9	8	c.1009G>C	rs137852253	306900
Hemophilia b	X	ChrX:138643869	F9	8	c.1025C>T	rs137852254	306900
Hemophilia b	X	ChrX:138643902	F9	8	c.1058T>C	rs137852255	306900
Hemophilia b	X	ChrX:138643908	F9	8	c.1064G>T	rs137852256	306900
Hemophilia b	X	ChrX:138643913	F9	8	c.1069G>A	rs137852257	306900
Hemophilia b	X	ChrX:138643914	F9	8	c.1070G>A	rs137852275	306900
Hemophilia b	X	ChrX:138643964	F9	8	c.1120G>T	rs137852271	306900
Hemophilia b	X	ChrX:138643979	F9	8	c.1135C>T	rs137852258	306900
Hemophilia b	X	ChrX:138643980	F9	8	c.1136G>A	rs137852259	306900
Hemophilia b	X	ChrX:138643994	F9	8	c.1150C>T	rs137852261	306900
Hemophilia b	X	ChrX:138644024	F9	8	c.1180A>G	rs137852262	306900
Hemophilia b	X	ChrX:138644031	F9	8	c.1187G>C	rs137852273	306900
Hemophilia b	X	ChrX:138644033	F9	8	c.1189G>C	rs137852281	306900
Hemophilia b	X	ChrX:138644076	F9	8	c.1232G>T	rs137852276	306900
Hemophilia b	X	ChrX:138644168	F9	8	c.1324G>A	rs137852267	306900
Hemophilia b	X	ChrX:138644172	F9	8	c.1328T>C	rs137852268	306900
Hemophilia b	X	ChrX:138644201	F9	8	c.1357T>C	rs137852269	306900
Hemophilia b	X	ChrX:138644213	F9	8	c.1369A>T	rs137852270	306900
Hereditary leiomyomatosis and renal cell cancer	1	Chr1:241676979	FH	3	c.302G>A	rs75086406	605839
Hurler syndrome	4	Chr4:981646	IDUA	2	c.208C>T	rs121965020	607014
Hurler syndrome	4	Chr4:996535	IDUA	9	c.1205G>A	rs121965019	607014
Hyperbilirubinemia, rotor type	12	Chr12:21349909	SLCO1B1	8	c.757C>T	rs183501729	237450
Hyperbilirubinemia, rotor type	12	Chr12:21375289	SLCO1B1	13	c.1738C>T	rs71581941	237450
Hypercholesterolemia, familial	19	Chr19:11216275	LDLR	4	c.693C>A	rs121908035	144400
Hypercholesterolemia, familial	19	Chr19:11218160	LDLR	6	c.910G>A	rs121908030	144400
Hypercholesterolemia, familial	19	Chr19:11231112	LDLR	14	c.2054C>T	rs28942084	144400
Hypercholesterolemia, familial	2	Chr2:21229160	APOB	26	c.10580G>A	rs5742904	144400
Hyperekplexia, hereditary 1	5	Chr5:151208511	GLRA1	8	c.1030C>T	rs281864913	149400
Hyperekplexia, hereditary 1	5	Chr5:151208570	GLRA1	8	c.971C>A	rs121918418	149400
Hyperinsulinemic hypoglycemia, familial, 2	11	Chr11:17408863	KCNJ11	1	c.776A>G	rs104894248	601820
Hyperinsulinemic hypoglycemia, familial, 4	4	Chr4:108948913	HADH	6	c.706C>T	rs375717077	609975
Hyperphenylalaninemia, bh4-deficient, d	10	Chr10:72643730	PCBD1	4	c.292C>T	rs121913015	264070
Hyperthyroxinemia, familial dysalbuminemic	4	Chr4:74277724	ALB	7	c.725G>A	rs75002628	615999

Hypogonadotropic hypogonadism 11 with or without anosmia	4	Chr4:104577415	TACR3	3	c.824G>A	rs144292455	614840
Hypogonadotropic hypogonadism 4 with or without anosmia	3	Chr3:71830623	PROK2	2	c.217C>T	rs121434272	610628
Hypogonadotropic hypogonadism 6 with or without anosmia	10	Chr10:103534966	FGF8	3	c.77C>T	rs137852660	612702
Hypomagnesemia 3, renal	3	Chr3:190122576	CLDN16	3	c.453G>T	rs104893729	248250
Hypophosphatasia, infantile	1	Chr1:21889712	ALPL	5	c.407G>A	rs121918011	241510
Hypophosphatasia, infantile	1	Chr1:21890632	ALPL	6	c.571G>A	rs121918007	241500
Hypophosphatasia, infantile	1	Chr1:21896819	ALPL	8	c.814C>T	rs121918020	241510
Hypophosphatasia, infantile	1	Chr1:21900176	ALPL	9	c.881A>C	rs121918002	241500
Hypophosphatasia, infantile	1	Chr1:21900274	ALPL	9	c.979T>C	rs121918010	241500
Hypophosphatasia, infantile	1	Chr1:21902229	ALPL	10	c.1001G>A	rs121918009	241500
Hypophosphatasia, infantile	1	Chr1:21902361	ALPL	10	c.1133A>T	rs121918008	241500
Hypothyroidism, congenital, nongoitrous, 1	14	Chr14:81610039	TSHR	10	c.1637G>A	rs121908866	275200
Hypouricemia, renal, 1	11	Chr11:64361219	SLC22A12	4	c.774G>A	rs121907892	220150
Ichthyosis prematurity syndrome	9	Chr9:131107776	SLC27A4	3	c.504C>A	rs137853131	608649
Ichthyosis, congenital, autosomal recessive 3	17	Chr17:8015495	ALOXE3	7	c.1096C>T	rs121434233	606545
Immunodeficiency, common variable, 2	17	Chr17:16852187	TNFRSF13	3	c.310T>C	rs34557412	240500
Immunoglobulin a deficiency 2	17	Chr17:16852187	TNFRSF13	3	c.310T>C	rs34557412	609529
Inclusion body myopathy 2, autosomal recessive	9	Chr9:36217445	GNE	12	c.2179G>T	rs121908627	600737
Inclusion body myopathy 2, autosomal recessive	9	Chr9:36218222	GNE	11	c.1984G>A	rs121908626	600737
Inclusion body myopathy 2, autosomal recessive	9	Chr9:36233990	GNE	5	c.1002T>A	rs121908628	600737
Irak4 deficiency	12	Chr12:44172041	IRAK4	9	c.877C>T	rs121908002	607676
Iron-refractory iron deficiency anemia	22	Chr22:37480815	TMPRSS6	9	c.1038C>A	rs137853121	206200
Joubert syndrome 2	11	Chr11:61161437	TMEM216	3	c.218G>T	rs201108965	608091
Joubert syndrome 3	6	Chr6:135754257	AHI1	14	c.2174G>A	-	608629
Joubert syndrome 3	6	Chr6:135768160	AHI1	11	c.1765C>T	rs267606641	608629
Joubert syndrome 5	12	Chr12:88471040	CEP290	41	c.5668G>T	rs137852832	610188
Joubert syndrome 5	12	Chr12:88477713	CEP290	36	c.4723A>T	rs137852834	610188
Joubert syndrome 6	8	Chr8:94767272	TMEM67	1	c.130C>T	rs267607118	610688
Joubert syndrome 6	8	Chr8:94777845	TMEM67	6	c.622A>T	rs137853108	610688
Joubert syndrome 7	16	Chr16:53686549	RPGRIP1L	15	c.2050C>T	rs121918204	611560
Joubert syndrome 7	16	Chr16:53720424	RPGRIP1L	6	c.697A>T	rs121918197	611560
Kanzaki disease	22	Chr22:42457043	NAGA	8	c.986G>A	rs121434533	609242
Lactase deficiency, congenital	2	Chr2:136564701	LCT	9	c.4170T>A	rs121908936	223000
Laron syndrome	5	Chr5:42629171	GHR	3	c.123G>A	rs121909370	262500
Laron syndrome	5	Chr5:42689023	GHR	4	c.189C>A	rs121909359	262500
Laron syndrome	5	Chr5:42689036	GHR	4	c.202C>T	rs121909358	262500
Laron syndrome	5	Chr5:42695055	GHR	5	c.324C>A	rs121909371	262500
Laron syndrome	5	Chr5:42711393	GHR	7	c.724C>T	rs121909363	262500
Leber congenital amaurosis 10	12	Chr12:88471040	CEP290	41	c.5668G>T	rs137852832	611755
Leber congenital amaurosis 10	12	Chr12:88477713	CEP290	36	c.4723A>T	rs137852834	611755
Leber congenital amaurosis 10	12	Chr12:88505097	CEP290	22	c.2249T>G	rs137852833	611755
Leber congenital amaurosis 13	14	Chr14:68191305	RDH12	4	c.184C>T	rs104894471	612712
Leber congenital amaurosis 2	1	Chr1:68903896	RPE65	10	c.1102T>C	rs62653011	204100
Leber congenital amaurosis 2	1	Chr1:68904716	RPE65	9	c.907A>T	rs61752904	204100

Leber congenital amaurosis 2	1	Chr1:68905269	RPE65	7	c.700C>T	rs61752895	204100
Leber congenital amaurosis 3	14	Chr14:88883069	SPATA7	5	c.253C>T	rs140287375	604232
Leber congenital amaurosis 3	14	Chr14:88883138	SPATA7	5	c.322C>T	rs80044281	604232
Leber congenital amaurosis 4	17	Chr17:6329101	AIPL1	6	c.834G>A	rs62637014	604393
Leber congenital amaurosis 5	6	Chr6:80203353	LCA5	4	c.835C>T	rs121918165	604537
Leber congenital amaurosis 8	1	Chr1:197390534	CRB1	6	c.1576C>T	rs114342808	613835
Lecithin:cholesterol acyltransferase deficiency	16	Chr16:67976870	LCAT	3	c.321C>A	rs121908055	245900
Left ventricular noncompaction 7	18	Chr18:19395685	MIB1	11	c.1588C>T	rs201850378	615092
Lesch-nyhan syndrome	X	ChrX:133607495	HPRT1	2	c.134G>A	rs137852491	300322
Lesch-nyhan syndrome	X	ChrX:133609285	HPRT1	3	c.209G>A	rs137852487	300322
Lesch-nyhan syndrome	X	ChrX:133609287	HPRT1	3	c.211G>C	rs137852488	300322
Lesch-nyhan syndrome	X	ChrX:133609298	HPRT1	3	c.222C>A	rs137852481	300322
Lesch-nyhan syndrome	X	ChrX:133627554	HPRT1	6	c.419G>A	rs137852503	300322
Lesch-nyhan syndrome	X	ChrX:133627594	HPRT1	6	c.459T>G	rs137852505	300322
Lesch-nyhan syndrome	X	ChrX:133632461	HPRT1	7	c.527C>T	rs137852493	300322
Lesch-nyhan syndrome	X	ChrX:133632463	HPRT1	7	c.529G>T	rs137852492	300322
Lesch-nyhan syndrome	X	ChrX:133632685	HPRT1	8	c.580G>A	rs267606863	300322
Lesch-nyhan syndrome	X	ChrX:133632700	HPRT1	8	c.595T>G	rs137852486	300322
Lesch-nyhan syndrome	X	ChrX:133634060	HPRT1	9	c.610C>G	rs137852490	300322
Lig4 syndrome	13	Chr13:108861177	LIG4	2	c.2440C>T	rs104894419	606593
Lipodystrophy, congenital generalized, type 3	7	Chr7:116166660	CAV1	2	c.112G>T	rs121434501	612526
Lipoid proteinosis of urbach and wiethe	1	Chr1:150482172	ECM1	3	c.157C>T	rs121909115	247100
Long qt syndrome 1	11	Chr11:2594101	KCNQ1	6	c.806G>A	rs120074194	192500
Long qt syndrome 1	11	Chr11:2594112	KCNQ1	6	c.817C>T	rs120074180	192500
Long qt syndrome 1	11	Chr11:2606443	KCNQ1	8	c.1034G>A	rs120074183	192500
Long qt syndrome 1	11	Chr11:2799220	KCNQ1	15	c.1747C>T	rs17221854	192500
Macular degeneration, juvenile	8	Chr8:87656008	CNGB3	10	c.1148delC	-	248200
Maple syrup urine disease	16	Chr16:31122037	BCKDK	8	c.671G>A	rs147210405	614923
Maple syrup urine disease	19	Chr19:41930487	BCKDHA	9	c.1312T>A	rs137852870	248600
Maple syrup urine disease	6	Chr6:80878662	BCKDHB	5	c.548G>C	rs79761867	248600
Meckel syndrome, type 1	4	Chr4:15504502	CC2D2A	7	c.394C>T	rs377177061	249000
Meckel syndrome, type 3	8	Chr8:94777845	TMEM67	6	c.622A>T	rs137853108	607361
Meckel syndrome, type 3	8	Chr8:94794684	TMEM67	11	c.1127A>C	rs137853106	607361
Meckel syndrome, type 5	16	Chr16:53679606	RPGRIP1L	17	c.2614C>T	rs121918203	611561
Methylmalonic aciduria and homocystinuria, cblc type	1	Chr1:45973938	MMACHC	3	c.331C>T	rs121918242	277400
Methylmalonic aciduria and homocystinuria, cblc type	1	Chr1:45974001	MMACHC	3	c.394C>T	rs121918241	277400
Methylmalonic aciduria and homocystinuria, cblc type	1	Chr1:45974520	MMACHC	4	c.482G>A	rs121918243	277400
Methylmalonyl-coa epimerase deficiency	2	Chr2:71351575	MCEE	2	c.139C>T	rs111033538	251120
Microcephalic osteodysplastic primordial dwarfism, type ii	21	Chr21:47773029	PCNT	10	c.1468C>T	rs181690344	210720
Microcephaly 6, primary, autosomal recessive	13	Chr13:25480590	CENPJ	7	c.1586C>G	rs202058504	608393
Microcephaly 9, primary, autosomal recessive	15	Chr15:49059645	CEP152	16	c.2034T>G	rs182018947	614852
Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma	14	Chr14:75022332	LTBP2	4	c.895C>T	rs121918355	251750
Mitochondrial complex i deficiency	2	Chr2:201950249	NDUFB3	4	c.208G>T	rs200800978	252010
Mitochondrial complex iii	2	Chr2:219525876	BCS1L	2	c.166C>T	rs121908576	124000

deficiency, nuclear type 1							
Mitochondrial complex iii deficiency, nuclear type 1	2	Chr2:219526568	BCS1L	4	c.547C>T	rs144885874	124000
Molybdenum cofactor deficiency, complementation group a	6	Chr6:39895101	MOCS1	2	c.217C>T	rs104893970	252150
Mononeuropathy of the median nerve, mild	5	Chr5:148406435	SH3TC2	11	c.2860C>T	rs80338933	613353
Mucolipidosis ii alpha/beta	12	Chr12:102147187	GNPTAB	19	c.3565C>T	rs137852897	252600
Mucolipidosis ii alpha/beta	12	Chr12:102147187	GNPTAB	19	c.3565C>T	rs137852897	252500
Mucolipidosis ii alpha/beta	12	Chr12:102147247	GNPTAB	19	c.3503_3504 d	rs34002892	252600
Mucolipidosis ii alpha/beta	12	Chr12:102147247	GNPTAB	19	c.3503_3504 d	rs34002892	252500
Mucolipidosis ii alpha/beta	12	Chr12:102151014	GNPTAB	18	c.3410T>A	rs142065232	252500
Mucolipidosis ii alpha/beta	12	Chr12:102164207	GNPTAB	9	c.1090C>T	rs200646278	252500
Mucolipidosis ii alpha/beta	12	Chr12:102183729	GNPTAB	3	c.310C>T	rs137852896	252500
Mucopolysaccharidosis, type iiia	12	Chr12:102164875	GNPTAB	8	c.832C>T	rs35878526	252900
Mucopolysaccharidosis, type iiia	17	Chr17:78188471	SGSH	4	c.449G>A	rs104894638	252900
Mucopolysaccharidosis, type iiia	17	Chr17:78190860	SGSH	2	c.220C>T	rs104894636	252900
Muscular dystrophy, congenital merosin-deficient, 1a	6	Chr6:129571327	LAMA2	13	c.1854_1861 d	rs202247791	607855
Muscular dystrophy, congenital merosin-deficient, 1a	6	Chr6:129573388	LAMA2	14	c.2049_2050 d	rs202247790	607855
Muscular dystrophy, congenital merosin-deficient, 1a	6	Chr6:129618874	LAMA2	21	c.2901C>A	rs121913577	607855
Muscular dystrophy, congenital merosin-deficient, 1a	6	Chr6:129636783	LAMA2	25	c.3718C>T	rs121913569	607855
Muscular dystrophy, congenital merosin-deficient, 1a	6	Chr6:129802567	LAMA2	55	c.7732C>T	rs121913572	607855
Muscular dystrophy, congenital merosin-deficient, 1a	6	Chr6:129837376	LAMA2	65	c.9253C>T	rs121913571	607855
Muscular dystrophy, limb-girdle, type 2a	15	Chr15:42676699	CAPN3	2	c.328C>T	rs121434545	253600
Muscular dystrophy, limb-girdle, type 2a	15	Chr15:42680000	CAPN3	4	c.550delA	rs80338800	253600
Muscular dystrophy, limb-girdle, type 2a	15	Chr15:42693953	CAPN3	11	c.1469G>A	rs121434548	253600
Muscular dystrophy, limb-girdle, type 2a	15	Chr15:42695170	CAPN3	13	c.1715G>A	rs121434544	253600
Muscular dystrophy, limb-girdle, type 2a	15	Chr15:42698129	CAPN3	15	c.1795dupA	rs745989418	253600
Muscular dystrophy, limb-girdle, type 2a	15	Chr15:42703124	CAPN3	22	c.2306G>A	rs80338802	253600
Muscular dystrophy, limb-girdle, type 2b	2	Chr2:71780261	DYSF	20	c.1927G>A	rs121908960	253601
Muscular dystrophy, limb-girdle, type 2b	2	Chr2:71901372	DYSF	52	c.5830C>T	rs121908959	253601
Myoclonic epilepsy of unverricht and lundborg	21	Chr21:45194178	CSTB	3	c.202C>T	rs74315442	254800
Myopathy, congenital, with fiber-type disproportion	1	Chr1:26136244	SEPN1	6	c.841G>A	rs121908188	255310
Nanophthalmos 2	11	Chr11:119216248	MFRP	5	c.523C>T	rs121908189	609549
Nephronophthisis 2	9	Chr9:103046624	INVS	13	c.1807C>T	rs121964994	602088
Nephronophthisis 2	9	Chr9:103055234	INVS	14	c.2695C>T	rs200844390	602088
Nephronophthisis 2	9	Chr9:103055258	INVS	14	c.2719C>T	rs267607185	602088
Neurodegeneration due to cerebral folate transport deficiency	11	Chr11:71906498	FOLR1	3	c.352C>T	rs121918405	613068
Neurodegeneration with brain iron accumulation 1	20	Chr20:3888734	PANK2	2	c.790C>T	rs137852961	234200
Neuropathy, hereditary sensory and autonomic, type iii	9	Chr9:111662583	IKBKAP	19	c.2087G>A	rs137853022	223900
Neutropenia, severe congenital, 4, autosomal recessive	17	Chr17:42153148	G6PC3	6	c.778G>C	rs200478425	612541
Niemann-pick disease, type a	11	Chr11:6413083	SMPD1	2	c.788T>A	rs120074120	257200

Niemann-pick disease, type a	11	Chr11:6415434	SMPD1	6	c.1493G>T	rs120074117	257200
Niemann-pick disease, type c1	18	Chr18:21118528	NPC1	20	c.3019C>G	rs80358257	257220
Niemann-pick disease, type c1	18	Chr18:21119357	NPC1	19	c.2873G>A	rs120074132	257220
Night blindness, congenital stationary, type 1b	5	Chr5:178413394	GRM6	8	c.1861C>T	rs62638214	257270
Night blindness, congenital stationary, type 1f	4	Chr4:110791223	LRIT3	4	c.1318C>T	-	615058
Nonaka myopathy	9	Chr9:36218221	GNE	11	c.1985C>T	rs62541771	605820
Obesity	18	Chr18:58038687	MC4R	1	c.896C>A	rs52804924	601665
Obesity	18	Chr18:58039478	MC4R	1	c.105C>A	rs13447324	601665
Odontoonychodermal dysplasia	2	Chr2:219747090	WNT10A	2	c.321C>A	rs121908119	257980
Orofaciodigital syndrome iv	10	Chr10:97442533	TCTN3	12	c.1327C>T	-	258860
Osteogenesis imperfecta, type viii	1	Chr1:43215921	LEPRE1	11	c.1656C>A	rs72659355	610915
Osteogenesis imperfecta, type viii	3	Chr3:33171463	CRTAP	4	c.826C>T	rs72659361	610854
Osteogenic sarcoma	17	Chr17:7577094	TP53	8	c.844C>T	rs28934574	259500
Osteogenic sarcoma	17	Chr17:7577548	TP53	7	c.733G>A	rs28934575	259500
Osteogenic sarcoma	22	Chr22:29130456	CHEK2	2	c.254C>T	rs17883862	259500
Osteopetrosis, autosomal recessive 1	11	Chr11:67814947	TCIRG1	11	c.1213G>A	rs137853150	259700
Osteopetrosis, autosomal recessive 1	11	Chr11:67815200	TCIRG1	12	c.1392C>A	rs137853149	259700
Pancreatitis, hereditary	1	Chr1:15767020	CTRC	3	c.164G>A	rs121909294	167800
Pancreatitis, hereditary	1	Chr1:15772212	CTRC	7	c.760C>T	rs121909293	167800
Pancreatitis, hereditary	5	Chr5:147211113	SPINK1	2	c.27delC	rs193922659	167800
Pancreatitis, hereditary	7	Chr7:117227832	CFTR	12	c.1624G>T	rs113993959	167800
Pancreatitis, hereditary	7	Chr7:117227860	CFTR	12	c.1652G>A	rs75527207	167800
Papillon-lefevre syndrome	11	Chr11:88027665	CTSC	7	c.901G>A	rs104894214	245000
Papillon-lefevre syndrome	11	Chr11:88029334	CTSC	6	c.856C>T	rs104894209	245000
Papillon-lefevre syndrome	11	Chr11:88042344	CTSC	4	c.628C>T	rs104894206	245000
Paragangliomas 5	5	Chr5:223624	SDHA	2	c.91C>T	rs142441643	614165
Parkinson disease 2, autosomal recessive juvenile	6	Chr6:162394349	PARK2	6	c.719C>T	rs137853054	600116
Pendred syndrome	7	Chr7:107302088	SLC26A4	2	c.2T>C	rs111033302	274600
Pendred syndrome	7	Chr7:107312690	SLC26A4	4	c.412G>T	rs111033199	274600
Pendred syndrome	7	Chr7:107315415	SLC26A4	6	c.626G>T	rs111033303	274600
Pendred syndrome	7	Chr7:107329499	SLC26A4	9	c.1003T>C	rs111033212	274600
Pendred syndrome	7	Chr7:107336429	SLC26A4	13	c.1489G>A	rs111033308	274600
Pendred syndrome	7	Chr7:107338530	SLC26A4	14	c.1588T>C	rs111033254	274600
Pendred syndrome	7	Chr7:107342468	SLC26A4	17	c.2000T>G	rs121908360	274600
Pendred syndrome	7	Chr7:107342483	SLC26A4	17	c.2015G>A	rs111033309	274600
Pendred syndrome	7	Chr7:107350577	SLC26A4	19	c.2168A>G	rs121908362	274600
Peripheral neuropathy, myopathy, hoarseness, and hearing loss	19	Chr19:50771512	MYH14	24	c.2921G>A	rs113993956	614369
Peroxisome biogenesis disorder 5a (zellweger)	8	Chr8:77896060	PEX2	4	c.355C>T	rs61752123	614866
Peroxisome biogenesis disorder 9b	6	Chr6:137166758	PEX7	4	c.345T>G	rs121909154	614879
Peters anomaly	2	Chr2:38302361	CYP1B1	2	c.171G>A	rs72549387	604229
Peutz-jeghers syndrome	19	Chr19:1221236	STK11	6	c.759C>A	rs137853075	175200
Phenylketonuria	12	Chr12:103234255	PAH	12	c.1238G>C	rs79931499	261600
Phenylketonuria	12	Chr12:103234270	PAH	12	c.1223G>A	rs5030859	261600
Phenylketonuria	12	Chr12:103234271	PAH	12	c.1222C>T	rs5030858	261600
Phenylketonuria	12	Chr12:103237466	PAH	11	c.1157A>G	rs62516141	261600

Phenylketonuria	12	Chr12:103237547	PAH	11	c.1076C>G	rs5030854	261600
Phenylketonuria	12	Chr12:103240716	PAH	9	c.926C>T	rs62642935	261600
Phenylketonuria	12	Chr12:103245479	PAH	8	c.898G>T	rs5030853	261600
Phenylketonuria	12	Chr12:103246653	PAH	7	c.782G>A	rs5030849	261600
Phenylketonuria	12	Chr12:103246659	PAH	7	c.776C>T	rs118203921	261600
Phenylketonuria	12	Chr12:103246671	PAH	7	c.764T>C	rs62642930	261600
Phenylketonuria	12	Chr12:103246714	PAH	7	c.721C>T	rs76687508	261600
Phenylketonuria	12	Chr12:103248926	PAH	6	c.694C>T	rs62507348	261600
Phenylketonuria	12	Chr12:103249059	PAH	6	c.561G>A	rs62507336	261600
Phenylketonuria	12	Chr12:103260410	PAH	5	c.473G>A	rs5030843	261600
Phenylketonuria	12	Chr12:103288534	PAH	3	c.331C>T	rs76296470	261600
Phenylketonuria	12	Chr12:103310906	PAH	1	c.3G>A	rs62514893	261600
Pigmented nodular adrenocortical disease, primary, 2	2	Chr2:178879181	PDE11A	2	c.919C>T	rs76308115	610475
Polycystic kidney disease, autosomal recessive	6	Chr6:51712669	PKHD1	50	c.8011C>T	rs137852947	263200
Polycystic kidney disease, autosomal recessive	6	Chr6:51923147	PKHD1	16	c.1486C>T	rs137852949	263200
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	6	Chr6:41129159	TREM2	2	c.233G>A	rs104893998	221770
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy	6	Chr6:41129295	TREM2	2	c.97C>T	rs104894002	221770
Prekallikrein deficiency	4	Chr4:187157943	KLKB1	5	c.337C>T	rs121964949	612423
Propionic acidemia	13	Chr13:100925472	PCCA	12	c.937C>T	rs138149179	606054
Prostate cancer, hereditary, 1	1	Chr1:182555149	RNASEL	2	c.793G>T	rs74315364	601518
Prostate cancer, hereditary, 1	1	Chr1:182555939	RNASEL	2	c.3G>A	rs74315365	601518
Pseudohypoaldosteronism, type i, autosomal recessive	12	Chr12:6458147	SCNN1A	10	c.1699C>T	rs137852634	264350
Pulmonary hypertension, primary, 1	2	Chr2:203378530	BMPR2	4	c.507C>A	rs137852747	178600
Pulmonary hypertension, primary, 1	2	Chr2:203421005	BMPR2	12	c.2617C>T	rs137852748	178600
Pulmonary hypertension, primary, 1	2	Chr2:203421084	BMPR2	12	c.2696G>A	rs137852752	178600
Pycnodysostosis	1	Chr1:150772083	CTSK	6	c.721C>T	rs74315303	265800
Pycnodysostosis	1	Chr1:150778585	CTSK	3	c.236G>A	rs74315305	265800
Pycnodysostosis	1	Chr1:150778667	CTSK	3	c.154A>T	rs74315306	265800
Pyridoxamine 5-prime-phosphate oxidase deficiency	17	Chr17:46024047	PNPO	7	c.685C>T	rs104894629	610090
Pyruvate kinase deficiency of red cells	1	Chr1:155261709	PKLR	10	c.1456C>T	rs116100695	266200
Retinitis pigmentosa 14	6	Chr6:35467877	TULP1	14	c.1376T>C	rs121909075	600132
Retinitis pigmentosa 20	1	Chr1:68903896	RPE65	10	c.1102T>C	rs62653011	613794
Retinitis pigmentosa 20	1	Chr1:68910541	RPE65	4	c.271C>T	rs61752871	613794
Retinitis pigmentosa 26	2	Chr2:182423344	CERKL	6	c.847C>T	rs121909398	608380
Retinitis pigmentosa 28	2	Chr2:62066830	FAM161A	3	c.1309A>T	rs200691042	606068
Retinitis pigmentosa 28	2	Chr2:62067454	FAM161A	3	c.685C>T	rs267606794	606068
Rhizomelic chondrodysplasia punctata, type 1	6	Chr6:137191088	PEX7	7	c.694C>T	rs121909153	215100
Rhizomelic chondrodysplasia punctata, type 1	6	Chr6:137219351	PEX7	9	c.875T>A	rs1805137	215100
Rigid spine muscular dystrophy 1	1	Chr1:26135587	SEPN1	5	c.716G>A	rs121908182	602771
Rigid spine muscular dystrophy 1	1	Chr1:26136244	SEPN1	6	c.841G>A	rs121908188	602771
Rothmund-thomson syndrome	8	Chr8:145738796	RECQL4	14	c.2269C>T	rs137853229	268400
Rubinstein-taybi syndrome 1	16	Chr16:3900690	CREBBP	2	c.406C>T	rs121434624	180849
Sarcosinemia	9	Chr9:136536816	SARDH	18	c.2167C>T	rs149391396	268900

Sarcosinemia	9	Chr9:136570084	SARDH	12	c.1540C>T	rs140559739	268900
Schopf-schulz-passarge syndrome	2	Chr2:219747090	WNT10A	2	c.321C>A	rs121908119	224750
Senior-loken syndrome 4	1	Chr1:5947496	NPHP4	18	c.2335C>T	rs137852922	606996
Senior-loken syndrome 4	1	Chr1:5964848	NPHP4	16	c.1972C>T	rs137852923	606996
Senior-loken syndrome 5	3	Chr3:121500619	IQCB1	13	c.1381C>T	rs121918244	609254
Senior-loken syndrome 5	3	Chr3:121508980	IQCB1	11	c.1069C>T	rs121918245	609254
Severe combined immunodeficiency with sensitivity to ionizing radiation	10	Chr10:14987109	DCLRE1C	3	c.241C>T	rs121908156	602450
Sitosterolemia	2	Chr2:44050063	ABCG5	10	c.1336C>T	rs199689137	210250
Sitosterolemia	2	Chr2:44099233	ABCG8	7	c.1083G>A	rs137852987	210250
Sitosterolemia	2	Chr2:44100948	ABCG8	9	c.1234C>T	rs137852991	210250
Spastic paraplegia 54, autosomal recessive	8	Chr8:38109734	DDHD2	13	c.1546C>T	rs373856119	615033
Spastic paraplegia 5a	8	Chr8:65509470	CYP7B1	6	c.1250G>A	rs121908611	270800
Spastic paraplegia 5a	8	Chr8:65517310	CYP7B1	5	c.1162C>T	rs72554620	270800
Spastic paraplegia 5a	8	Chr8:65528273	CYP7B1	3	c.825T>A	rs121908613	270800
Spastic paraplegia 7, autosomal recessive	16	Chr16:89576947	SPG7	2	c.233T>A	rs121918358	607259
Spondylocostal dysostosis 1, autosomal recessive	19	Chr19:39994770	DLL3	5	c.712C>T	rs104894675	277300
Stuve-wiedemann syndrome	5	Chr5:38496580	LIFR	13	c.1789C>T	rs121912501	601559
Succinic semialdehyde dehydrogenase deficiency	6	Chr6:24528285	ALDH5A1	9	c.1273C>T	rs118203983	271980
Supranuclear palsy, progressive, 1	17	Chr17:44039717	MAPT	2	c.14G>A	rs63750959	601104
Tangier disease	9	Chr9:107593329	ABCA1	14	c.1769G>C	rs137854496	205400
Three m syndrome 1	6	Chr6:43006687	CUL7	23	c.4585C>T	rs121918228	273750
Thrombophilia due to protein c deficiency	2	Chr2:128183756	PROC	7	c.631C>T	rs121918143	176860
Thrombophilia due to protein c deficiency	2	Chr2:128183783	PROC	7	c.658C>T	rs121918152	176860
Thrombophilia due to protein c deficiency	2	Chr2:128183803	PROC	7	c.678G>C	rs121918155	176860
Thrombophilia due to protein c deficiency	2	Chr2:128186002	PROC	9	c.866C>T	rs121918151	612304
Thrombophilia due to protein c deficiency	2	Chr2:128186071	PROC	9	c.935C>T	rs121918160	176860
Thrombophilia due to protein c deficiency	2	Chr2:128186163	PROC	9	c.1027G>A	rs121918147	612304
Thrombophilia due to protein c deficiency	2	Chr2:128186471	PROC	9	c.1335C>G	rs121918157	612304
Thrombophilia due to protein c deficiency	3	Chr3:93617306	PROS1	8	c.835C>T	rs121918475	612336
Thrombotic thrombocytopenic purpura, congenital	9	Chr9:136319562	ADAMTS13	24	c.3070T>G	rs121908472	274150
Thyroid dyshormonogenesis 3	8	Chr8:133894854	TG	7	c.886C>T	rs121912648	274700
Thyroid dyshormonogenesis 3	8	Chr8:133935642	TG	22	c.4588C>T	rs121912646	274700
Tooth agenesis, selective, 4	2	Chr2:219747090	WNT10A	2	c.321C>A	rs121908119	150400
Treacher collins syndrome 3	6	Chr6:43488699	POLR1C	8	c.835C>T	rs141156009	248390
Treacher collins syndrome 3	6	Chr6:43488700	POLR1C	8	c.836G>A	rs191582628	248390
Trimethylaminuria	1	Chr1:171083232	FMO3	7	c.913G>T	rs61753344	602079
Tumoral calcinosis, hyperphosphatemic, familial	2	Chr2:166611525	GALNT3	8	c.1441C>T	rs137853089	610233
Tumoral calcinosis, hyperphosphatemic, familial	2	Chr2:166626727	GALNT3	2	c.484C>T	rs137853086	610233
Tyrosinemia, type i	15	Chr15:80465435	FAH	9	c.786G>A	rs80338899	276700
Tyrosinemia, type i	15	Chr15:80473390	FAH	13	c.1069G>T	rs121965075	276700
Tyrosinemia, type i	15	Chr15:80473411	FAH	13	c.1090G>T	rs121965076	276700
Usher syndrome, type i	11	Chr11:76867729	MYO7A	6	c.494C>T	rs111033174	276900
Usher syndrome, type i	11	Chr11:76867812	MYO7A	6	c.582delC	rs111033238	276900

Usher syndrome, type i	11	Chr11:76867950	MYO7A	7	c.635G>A	rs28934610	276900
Usher syndrome, type i	11	Chr11:76868015	MYO7A	7	c.700C>T	rs41298133	276900
Usher syndrome, type i	11	Chr11:76869472	MYO7A	9	c.999T>G	rs111033285	276900
Usher syndrome, type i	11	Chr11:76883896	MYO7A	16	c.1900C>T	rs111033180	276900
Usher syndrome, type i	11	Chr11:76885862	MYO7A	17	c.1996C>T	rs121965085	276900
Usher syndrome, type i	11	Chr11:76885871	MYO7A	17	c.2005C>T	rs111033201	276900
Usher syndrome, type i	11	Chr11:76894153	MYO7A	26	c.3327delC	rs111033433	276900
Usher syndrome, type i	11	Chr11:76901153	MYO7A	29	c.3719G>A	rs111033178	276900
Usher syndrome, type i	11	Chr11:76901753	MYO7A	30	c.3764delA	rs111033347	276900
Usher syndrome, type i	11	Chr11:76913402	MYO7A	37	c.5101C>T	rs111033182	276900
Usher syndrome, type i	11	Chr11:76919517	MYO7A	43	c.5899C>T	rs376764423	276900
Usher syndrome, type i	11	Chr11:76919826	MYO7A	44	c.6029A>G	rs111033175	276900
Usher syndrome, type i	11	Chr11:76922215	MYO7A	45	c.6070C>T	rs111033198	276900
Usher syndrome, type if	10	Chr10:55698632	PCDH15	26	c.3331C>T	rs202033121	602083
Usher syndrome, type if	10	Chr10:55849801	PCDH15	17	c.1955C>G	rs137853004	602083
Usher syndrome, type if	10	Chr10:56077174	PCDH15	9	c.748C>T	rs111033260	602083
Usher syndrome, type if	10	Chr10:56424016	PCDH15	2	c.7C>T	rs137853001	602083
Usher syndrome, type iiia	3	Chr3:150645894	CLRN1	4	c.567T>G	rs121908140	276902
Usher syndrome, type iiia	3	Chr3:150659434	CLRN1	3	c.540C>A	rs374963432	276902
Usher syndrome, type iiia	3	Chr3:150690307	CLRN1	1	c.189C>A	rs111033267	276902
Van der woude syndrome 1	1	Chr1:209974625	IRF6	3	c.134G>A	rs121434229	119300
Vas deferens, congenital bilateral aplasia of	7	Chr7:117304834	CFTR	25	c.4056G>C	rs113857788	277180
Vitamin d hydroxylation-deficient rickets, type 1a	12	Chr12:58157930	CYP27B1	7	c.1166G>A	rs118204009	264700
Vitamin d hydroxylation-deficient rickets, type 1a	12	Chr12:58157931	CYP27B1	7	c.1165C>G	rs118204010	264700
Vitamin d hydroxylation-deficient rickets, type 1a	12	Chr12:58159103	CYP27B1	3	c.566A>G	rs118204012	264700
Von willebrand disease, type 1	12	Chr12:6122710	VWF	32	c.5557C>T	rs61750612	193400
Von willebrand disease, type 1	12	Chr12:6127609	VWF	28	c.4975C>T	rs61750595	193400
Von willebrand disease, type 1	12	Chr12:6143978	VWF	20	c.2561G>A	rs41276738	193400
Von willebrand disease, type 1	12	Chr12:6155891	VWF	17	c.2279G>A	rs61748467	193400
Werner syndrome	8	Chr8:30938648	WRN	9	c.1105C>T	rs17847577	277700
Werner syndrome	8	Chr8:31004913	WRN	30	c.3493C>T	rs121908447	277700
Werner syndrome	8	Chr8:31014977	WRN	33	c.3913C>T	rs121908446	277700
Wilson disease	13	Chr13:52511478	ATP7B	19	c.3955C>T	rs193922109	277900
Wilson disease	13	Chr13:52511706	ATP7B	18	c.3809A>G	rs121907990	277900
Wilson disease	13	Chr13:52511719	ATP7B	18	c.3796G>A	rs121907992	277900
Wilson disease	13	Chr13:52518281	ATP7B	14	c.3207C>A	rs76151636	277900
Wilson disease	13	Chr13:52520550	ATP7B	13	c.2930C>T	rs72552255	277900
Wilson disease	13	Chr13:52520574	ATP7B	13	c.2906G>A	rs121907996	277900
Wilson disease	13	Chr13:52523836	ATP7B	12	c.2827G>A	rs28942076	277900
Wilson disease	13	Chr13:52524252	ATP7B	11	c.2621C>T	rs121907994	277900
Wilson disease	13	Chr13:52524268	ATP7B	11	c.2605G>A	rs191312027	277900
Wilson disease	13	Chr13:52532466	ATP7B	8	c.2336G>A	rs137853283	277900
Wilson disease	13	Chr13:52532469	ATP7B	8	c.2333G>T	rs28942074	277900
Wilson disease	13	Chr13:52532470	ATP7B	8	c.2332C>T	rs137853284	277900
Wilson disease	13	Chr13:52532497	ATP7B	8	c.2305A>G	rs193922103	277900
Wilson disease	13	Chr13:52532505	ATP7B	8	c.2297C>G	rs121907997	277900
Wilson disease	13	Chr13:52548491	ATP7B	2	c.865C>T	rs121907999	277900

Wilson disease	13	Chr13:52548510	ATP7B	2	c.845delT	rs193922111	277900
Xeroderma pigmentosum, complementation group a	9	Chr9:100437861	XPA	6	c.682C>T	rs104894132	278700