Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
3-beta-hydroxysteroid dehydrogenase deficiency, type II	HSD3B2	AR	General Population	<500	99	<49901
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL	AR	General Population	<500	99	<49901
3-hydroxyacyl-CoA dehydrogenase deficiency	HADH	AR	General Population	<500	99	<49901
3-methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1	AR	General Population	147	99	14601
3-methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1	AR	Caucasian	137	99	13601
3-methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2	AR	General Population	120	99	11901
3-methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2	AR	Caucasian	112	99	11101
3-methylglutaconic aciduria, type III / Costeff syndrome	OPA3	AR	General Population	<500	99	<49901
3-methylglutaconic aciduria, type III / Costeff syndrome	OPA3	AR	Sephardic Jewish - Iraqi	13	99	1201
6-pyruvoyl-tetrahydropterin synthase deficiency	PTS	AR	General Population	<500	99	<49901
6-pyruvoyl-tetrahydropterin synthase deficiency	PTS	AR	East Asian	158	99	15701
6-pyruvoyl-tetrahydropterin synthase deficiency	PTS	AR	Asian	122	99	12101
Abetalipoproteinemia	MTTP	AR	Caucasian	<500	99	<49901
Abetalipoproteinemia	MTTP	AR	General Population	<500	99	<49901
Abetalipoproteinemia	MTTP	AR	Ashkenazi Jewish	186	99	18501
Achromatopsia, CNGB3-related	CNGB3	AR	General Population	98	99	9701
Achromatopsia, CNGB3-related	CNGB3	AR	Caucasian	91	99	9001
Acrodermatitis enteropathica	SLC39A4	AR	General Population	354	99	35301
Acute infantile liver failure	TRMU	AR	General Population	<500	99	<49901
Acute infantile liver failure	TRMU	AR	Sephardic Jewish - Yemenite	34	99	3301
Adenosine deaminase deficiency	ADA	AR	General Population	224	99	22301
Adrenoleukodystrophy, X-linked	ABCD1	XL	General Population	10500	99	1049901
Adrenoleukodystrophy, X-linked	ABCD1	XL	Sephardic Jewish	10500	99	1049901
Agenesis of the corpus callosum with peripheral neuropathy	SLC12A6	AR	General Population	<500	99	<49901
Agenesis of the corpus callosum with peripheral neuropathy	SLC12A6	AR	French Canadian	23	99	2201
Aicardi-Goutieres syndrome 5	SAMHD1	AR	General Population	<500	99	<49901
Alpha-1 antitrypsin deficiency	SERPINA1	AR	General Population	38	99	3701
Alpha-1 antitrypsin deficiency	SERPINA1	AR	Ashkenazi Jewish	24	99	2301
Alpha-1 antitrypsin deficiency	SERPINA1	AR	Northern European Caucasian	15	99	1401
Alpha-mannosidosis	MAN2B1	AR	General Population	<500	99	<49901
Alpha-mannosidosis	MAN2B1	AR	Caucasian	485	99	48401
Alpha-mannosidosis	MAN2B1	AR	Northern European Caucasian	354	99	35301
Alpha-thalassemia	HBA1/HBA2	AR	Caucasian	<500	95	<9981
Alpha-thalassemia	HBA1/HBA2	AR	African American	30	95	581
Alpha-thalassemia	HBA1/HBA2	AR	General Population	25	95	481
Alpha-thalassemia	HBA1/HBA2	AR	Asian	20	95	381
Alpha-thalassemia	HBA1/HBA2	AR	East Asian	16	95	301
Alpha-thalassemia	HBA1/HBA2	AR	Southeast Asian	7	95	121
Alpha-thalassemia	HBA1/HBA2	AR	South Asian	2	95	21
Alpha-thalassemia intellectual disability syndrome, X-linked	ATRX	XL	General Population	<750000	99	<74999901
Alport syndrome, COL4A3-related	COL4A3	AR	General Population	323	99	32201
Alport syndrome, COL4A3-related	COL4A3	AR	Caucasian	284	99	28301
Alport syndrome, COL4A3-related	COL4A3	AR	Ashkenazi Jewish	189	99	18801
Alport syndrome, COL4A4-related	COL4A4	AR	General Population	353	99	35201
Alport syndrome, COL4A5-related, X-linked	COL4A5	XL	General Population	47000	99	4699901
Alstrom syndrome	ALMS1	AR	General Population	<500	99	<49901
Argininemia	ARG1	AR	General Population	<500	99	<49901
Argininosuccinic aciduria	ASL	AR	Finnish	190	99	18901
Argininosuccinic aciduria	ASL	AR	General Population	133	99	13201
Aromatase deficiency	CYP19A1	AR	General Population	<500	99	<49901
Arthrogryposis, mental retardation, and seizures	SLC35A3	AR	General Population	<500	99	<49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Arthrogryposis, mental retardation, and seizures	SLC35A3	AR	Ashkenazi Jewish	453	99	45201
Asparagine synthetase deficiency	ASNS	AR	General Population	<500	99	<49901
Asparagine synthetase deficiency	ASNS	AR	Sephardic Jewish - Iranian	80	99	7901
Aspartylglycosaminuria	AGA	AR	General Population	<500	99	<49901
Aspartylglycosaminuria	AGA	AR	Caucasian	<500	99	<49901
Aspartylglycosaminuria	AGA	AR	Finnish	36	99	3501
Ataxia with isolated vitamin E deficiency	TTPA	AR	General Population	<500	99	<49901
Ataxia with isolated vitamin E deficiency	TTPA	AR	Caucasian	<500	99	<49901
Ataxia-telangiectasia	ATM	AR	Ashkenazi Jewish	<500	99	<49901
Ataxia-telangiectasia	ATM	AR	General Population	100	99	9901
Ataxia-telangiectasia	ATM	AR	Sephardic Jewish - Moroccan	81	99	8001
Autoimmune polyglandular syndrome, type 1	AIRE	AR	General Population	354	99	35301
Autoimmune polyglandular syndrome, type 1	AIRE	AR	Finnish	79	99	7801
Autoimmune polyglandular syndrome, type 1	AIRE	AR	Sardinian	60	99	5901
Autoimmune polyglandular syndrome, type 1	AIRE	AR	Sephardic Jewish - Iranian	27	99	2601
Autosomal recessive congenital ichthyosis	TGM1	AR	General Population	301	99	30001
Autosomal recessive congenital ichthyosis	TGM1	AR	Caucasian	253	99	25201
Autosomal recessive congenital ichthyosis	TGM1	AR	Norwegian	151	99	15001
Autosomal recessive polycystic kidney disease	PKHD1	AR	General Population	144	99	14301
Autosomal recessive polycystic kidney disease	PKHD1	AR	Ashkenazi Jewish	106	99	10501
Autosomal recessive polycystic kidney disease	PKHD1	AR	Caucasian	100	99	9901
Autosomal recessive polycystic kidney disease	PKHD1	AR	South African Afrikaner	52	99	5101
Bardet-Biedl syndrome 1	BBS1	AR	General Population	265	99	26401
Bardet-Biedl syndrome 1	BBS1	AR	Faroese	30	99	2901
Bardet-Biedl syndrome 10	BBS10	AR	General Population	447	99	44601
Bardet-Biedl syndrome 12	BBS12	AR	General Population	<500	99	<49901
Bardet-Biedl syndrome 2	BBS2	AR	General Population	<500	99	<49901
Bardet-Biedl syndrome 2	BBS2	AR	Ashkenazi Jewish	135	99	13401
Bardet-Biedl syndrome 2	BBS2	AR	Hutterites	22	99	2101
Bardet-Biedl syndrome 6	MKKS	AR	General Population	219	99	21801
Bare lymphocyte syndrome, type II	CIITA	AR	General Population	<500	99	<49901
Bartter syndrome, type 4A	BSND	AR	General Population	<500	99	<49901
Bernard-Soulier syndrome, type A	GP1BA	AR	General Population	<500	99	<49901
Bernard-Soulier syndrome, type C	GP9	AR	General Population	<500	99	<49901
Beta hemoglobinopathies	HBB	AR	Caucasian	373	99	37201
Beta hemoglobinopathies	HBB	AR	General Population	129	99	12801
Beta hemoglobinopathies	HBB	AR	Hispanic	83	99	8201
Beta hemoglobinopathies	HBB	AR	East Asian	78	99	7701
Beta hemoglobinopathies	HBB	AR	Southern European Caucasian	59	99	5801
Beta hemoglobinopathies	HBB	AR	Asian	54	99	5301
Beta hemoglobinopathies	HBB	AR	South Asian	32	99	3101
Beta hemoglobinopathies	HBB	AR	Southeast Asian	30	99	2901
Beta hemoglobinopathies	HBB	AR	Mediterranean	28	99	2701
Beta hemoglobinopathies	HBB	AR	African American	10	99	901
Beta hemoglobinopathies	HBB	AR	Middle Eastern	5	99	401
Beta-ketothiolase deficiency	ACAT1	AR	Caucasian	354	99	35301
Beta-ketothiolase deficiency	ACAT1	AR	General Population	347	99	34601
Beta-ketothiolase deficiency	ACAT1	AR	Asian	289	99	28801
Bilateral frontoparietal polymicrogyria	ADGRG1	AR	General Population	<500	99	<49901
Biotinidase deficiency	BTD	AR	General Population	120	99	11901
Biotinidase deficiency	BTD	AR	Hispanic	30	99	2901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Biotinidase deficiency	BTD	AR	Caucasian	12	99	1101
Bloom syndrome	BLM	AR	General Population	<500	99	<49901
Bloom syndrome	BLM	AR	Ashkenazi Jewish	140	99	13901
Canavan disease	ASPA	AR	General Population	<500	99	<49901
Canavan disease	ASPA	AR	Ashkenazi Jewish	60	99	5901
Carbamoyl phosphate synthetase I deficiency	CPS1	AR	General Population	<500	99	<49901
Carbamoyl phosphate synthetase I deficiency	CPS1	AR	Asian	447	99	44601
Carbamoyl phosphate synthetase I deficiency	CPS1	AR	Caucasian	284	99	28301
Carnitine deficiency, systemic primary	SLC22A5	AR	General Population	200	99	19901
Carnitine deficiency, systemic primary	SLC22A5	AR	Caucasian	110	99	10901
Carnitine deficiency, systemic primary	SLC22A5	AR	East Asian	100	99	9901
Carnitine deficiency, systemic primary	SLC22A5	AR	Asian	100	99	9901
Carnitine deficiency, systemic primary	SLC22A5	AR	Faroese	20	99	1901
Carnitine palmitoyltransferase I deficiency	CPT1A	AR	General Population	<500	99	<49901
Carnitine palmitoyltransferase I deficiency	CPT1A	AR	Hutterites	16	99	1501
Carnitine palmitoyltransferase II deficiency	CPT2	AR	General Population	<500	99	<49901
Carnitine palmitoyltransferase II deficiency	CPT2	AR	Asian	<500	99	<49901
Carnitine palmitoyltransferase II deficiency	CPT2	AR	African American	308	99	30701
Carnitine palmitoyltransferase II deficiency	CPT2	AR	Caucasian	200	99	19901
Carnitine palmitoyltransferase II deficiency	CPT2	AR	Ashkenazi Jewish	51	99	5001
Carnitine-acylcarnitine translocase deficiency	SLC25A20	AR	General Population	<500	99	<49901
Carpenter syndrome	RAB23	AR	General Population	<500	99	<49901
Carpenter syndrome	RAB23	AR	Caucasian	<500	99	<49901
Cartilage-hair hypoplasia	RMRP	AR	General Population	<500	99	<49901
Cartilage-hair hypoplasia	RMRP	AR	Finnish	76	99	7501
Cartilage-hair hypoplasia	RMRP	AR	Amish	19	99	1801
Cerebrooculofacioskeletal syndrome 1 / Cockayne syndrome, type B	ERCC6	AR	General Population	<500	99	<49901
Cerebrotendinous xanthomatosis	CYP27A1	AR	Southern European Caucasian	<500	99	<49901
Cerebrotendinous xanthomatosis	CYP27A1	AR	General Population	115	99	11401
Cerebrotendinous xanthomatosis	CYP27A1	AR	Sephardic Jewish - Moroccan	5	99	401
Charcot-Marie-Tooth disease, type 1X	GJB1	XL	General Population	7000	99	699901
Charcot-Marie-Tooth disease, type 4D	NDRG1	AR	General Population	<500	99	<49901
Charcot-Marie-Tooth disease, type 4D	NDRG1	AR	Roma	22	99	2101
Choreoacanthocytosis	VPS13A	AR	General Population	<500	99	<49901
Choreoacanthocytosis	VPS13A	AR	Ashkenazi Jewish	<500	99	<49901
Choroideremia, X-linked	CHM	XL	General Population	25000	99	2499901
Chronic granulomatous disease 4	CYBA	AR	General Population	<500	99	<49901
Chronic granulomatous disease 4	CYBA	AR	Sephardic Jewish - Moroccan	13	99	1201
Chronic granulomatous disease, X-linked	CYBB	XL	General Population	150000	99	14999901
Ciliopathies, RPGRIP1L-related	RPGRIP1L	AR	General Population	259	99	25801
Citrin deficiency / Citrullinemia, type II	SLC25A13	AR	Caucasian	<500	99	<49901
Citrin deficiency / Citrullinemia, type II	SLC25A13	AR	General Population	<500	99	<49901
Citrin deficiency / Citrullinemia, type II	SLC25A13	AR	Asian	123	99	12201
Citrin deficiency / Citrullinemia, type II	SLC25A13	AR	East Asian	65	99	6401
Citrullinemia, type I	ASS1	AR	Caucasian	195	99	19401
Citrullinemia, type I	ASS1	AR	Asian	123	99	12201
Citrullinemia, type I	ASS1	AR	General Population	119	99	11801
Cockayne syndrome, type A	ERCC8	AR	General Population	<500	99	<49901
Cohen syndrome	VPS13B	AR	General Population	<500	99	<49901
Combined malonic and methylmalonic aciduria	ACSF3	AR	General Population	86	99	8501
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	MMACHC	AR	General Population	138	99	13701

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	MMACHC	AR	Caucasian	138	99	13701
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	MMACHC	AR	Asian	113	99	11201
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	MMACHC	AR	East Asian	112	99	11101
Combined methylmalonic aciduria and homocystinuria, cblD type / Cobalamin D deficiency	MMADHC	AR	Caucasian	<500	99	<49901
Combined methylmalonic aciduria and homocystinuria, cblD type / Cobalamin D deficiency	MMADHC	AR	General Population	<500	99	<49901
Combined oxidative phosphorylation deficiency 1	GFM1	AR	General Population	<500	99	<49901
Combined oxidative phosphorylation deficiency 3	TSFM	AR	General Population	<500	99	<49901
Combined oxidative phosphorylation deficiency 3	TSFM	AR	Finnish	80	99	7901
Combined pituitary hormone deficiency, type 2	PROP1	AR	General Population	141	99	14001
Combined pituitary hormone deficiency, type 3	LHX3	AR	General Population	<500	99	<49901
Congenital adrenal hyperplasia (CAH) due to 11-beta-hydroxylase deficiency	CYP11B1	AR	General Population	158	99	15701
Congenital adrenal hyperplasia (CAH) due to 17-alpha-hydroxylase deficiency	CYP17A1	AR	General Population	<500	99	<49901
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	CYP21A2	AR	Caucasian	67	98	3301
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	CYP21A2	AR	General Population	61	98	3001
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	CYP21A2	AR	Ashkenazi Jewish	40	98	1951
Congenital amegakaryocytic thrombocytopenia	MPL	AR	General Population	415	99	41401
Congenital amegakaryocytic thrombocytopenia	MPL	AR	Caucasian	266	99	26501
Congenital amegakaryocytic thrombocytopenia	MPL	AR	Ashkenazi Jewish	57	99	5601
Congenital disorder of glycosylation, type Ia	PMM2	AR	Asian	449	99	44801
Congenital disorder of glycosylation, type Ia	PMM2	AR	General Population	124	99	12301
Congenital disorder of glycosylation, type Ia	PMM2	AR	Ashkenazi Jewish	61	99	6001
Congenital disorder of glycosylation, type Ia	PMM2	AR	Northern European Caucasian	60	99	5901
Congenital disorder of glycosylation, type Ia	PMM2	AR	Caucasian	42	99	4101
Congenital disorder of glycosylation, type Ib	MPI	AR	General Population	<500	99	<49901
Congenital disorder of glycosylation, type Ic	ALG6	AR	General Population	<500	99	<49901
Congenital insensitivity to pain with anhidrosis	NTRK1	AR	Sephardic Jewish - Moroccan	<500	99	<49901
Congenital insensitivity to pain with anhidrosis	NTRK1	AR	General Population	<500	99	<49901
Congenital insensitivity to pain with anhidrosis	NTRK1	AR	Asian	387	99	38601
Congenital myasthenic syndrome, CHRNE-related	CHRNE	AR	General Population	<500	99	<49901
Congenital myasthenic syndrome, CHRNE-related	CHRNE	AR	Caucasian	383	99	38201
Congenital myasthenic syndrome, RAPSN-related	RAPSN	AR	Sephardic Jewish - Iraqi, Iranian	<500	99	<49901
Congenital myasthenic syndrome, RAPSN-related	RAPSN	AR	General Population	252	99	25101
Congenital myasthenic syndrome, RAPSN-related	RAPSN	AR	Caucasian	176	99	17501
Congenital neutropenia, HAX1-related	HAX1	AR	General Population	<500	99	<49901
Corneal dystrophy and perceptive deafness syndrome	SLC4A11	AR	General Population	<500	99	<49901
Corticosterone methyloxidase deficiency	CYP11B2	AR	General Population	<500	99	<49901
Corticosterone methyloxidase deficiency	CYP11B2	AR	Sephardic Jewish - Iranian	30	99	2901
CRB1-related retinal dystrophies	CRB1	AR	General Population	112	99	11101
Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome	SLC6A8	XL	General Population	20600	99	2059901
Cystic fibrosis	CFTR	AR	South Asian	90	99	8901
Cystic fibrosis	CFTR	AR	Southeast Asian	90	99	8901
Cystic fibrosis	CFTR	AR	East Asian	90	99	8901
Cystic fibrosis	CFTR	AR	African American	61	99	6001
Cystic fibrosis	CFTR	AR	Hispanic	46	99	4501
Cystic fibrosis	CFTR	AR	Northern European Caucasian	25	99	2401
Cystic fibrosis	CFTR	AR	Ashkenazi Jewish	25	99	2401
Cystic fibrosis	CFTR	AR	Caucasian	25	99	2401
Cystic fibrosis	CFTR	AR	General Population	32	99	3101
Cystinosis	CTNS	AR	African American	<500	99	<49901
Cystinosis	CTNS	AR	Asian	<500	99	<49901
Cystinosis	CTNS	AR	Hispanic	<500	99	<49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Cystinosis	CTNS	AR	Caucasian	220	99	21901
Cystinosis	CTNS	AR	General Population	158	99	15701
Cystinosis	CTNS	AR	Sephardic Jewish - Moroccan	100	99	9901
Cystinosis	CTNS	AR	French Canadian - Saguenay Lac-St. Jean	39	99	3801
D-bifunctional protein deficiency	HSD17B4	AR	General Population	158	99	15701
Dihydrolipoamide dehydrogenase deficiency	DLD	AR	General Population	<500	99	<49901
Dihydrolipoamide dehydrogenase deficiency	DLD	AR	Ashkenazi Jewish	94	99	9301
Duchenne/Becker muscular dystrophy, X-linked	DMD	XL	General Population	4200	99	419901
Dyskeratosis congenita, RTEL1-related	RTEL1	AR	General Population	<500	99	<49901
Dyskeratosis congenita, RTEL1-related	RTEL1	AR	Ashkenazi Jewish	165	99	16401
Dystrophic epidermolysis bullosa, COL7A1-related	COL7A1	AR	General Population	370	99	36901
Ehlers-Danlos syndrome, dermatosparaxis type	ADAMTS2	AR	General Population	<500	99	<49901
Ehlers-Danlos syndrome, dermatosparaxis type	ADAMTS2	AR	Ashkenazi Jewish	248	99	24701
Ellis-van Creveld syndrome	EVC	AR	General Population	345	99	34401
Ellis-van Creveld syndrome	EVC	AR	Lancaster County Amish	12	99	1101
Ellis-van Creveld syndrome	EVC2	AR	General Population	122	99	12101
Emery-Dreifuss muscular dystrophy, X-linked	EMD	XL	General Population	250000	99	24999901
Enhanced S-cone syndrome	NR2E3	AR	General Population	204	99	20301
Enhanced S-cone syndrome	NR2E3	AR	Ashkenazi Jewish	100	99	9901
Ethylmalonic encephalopathy	ETHE1	AR	General Population	<500	99	<49901
Fabry disease, X-linked	GLA	XL	General Population	1500	99	149901
Factor IX deficiency / Hemophilia B	F9	XL	General Population	10000	99	999901
Factor XI deficiency / Hemophilia C	F11	AR	Asian	163	99	16201
Factor XI deficiency / Hemophilia C	F11	AR	Caucasian	101	99	10001
Factor XI deficiency / Hemophilia C	F11	AR	General Population	92	99	9101
Factor XI deficiency / Hemophilia C	F11	AR	Ashkenazi Jewish	11	99	1001
Familial dysautonomia	ELP1	AR	General Population	<500	99	<49901
Familial dysautonomia	ELP1	AR	Ashkenazi Jewish	34	99	3301
Familial hypercholesterolemia, LDLRAP1-related	LDLRAP1	AR	General Population	<500	99	<49901
Familial hypercholesterolemia, LDLRAP1-related	LDLRAP1	AR	Sardinian	143	99	14201
Familial hypercholesterolemia, LDLR-related	LDLR	AR	General Population	<500	99	<49901
Familial hypercholesterolemia, LDLR-related	LDLR	AR	French Canadian	267	99	26601
Familial hypercholesterolemia, LDLR-related	LDLR	AR	Caucasian	200	99	19901
Familial hypercholesterolemia, LDLR-related	LDLR	AR	Finnish	143	99	14201
Familial hypercholesterolemia, LDLR-related	LDLR	AR	South African Afrikaner	70	99	6901
Familial hypercholesterolemia, LDLR-related	LDLR	AR	Ashkenazi Jewish	69	99	6801
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	General Population	112	99	11101
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	Ashkenazi Jewish	52	99	5101
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	Finnish	29	99	2801
Familial hyperinsulinism, KCNJ11-related	KCNJ11	AR	General Population	<500	99	<49901
Familial Mediterranean fever	MEFV	AR	General Population	115	99	11401
Familial Mediterranean fever	MEFV	AR	Sephardic Jewish	14	99	1301
Familial Mediterranean fever	MEFV	AR	Ashkenazi Jewish	13	99	1201
Familial Mediterranean fever	MEFV	AR	Armenian	5	99	401
Familial Mediterranean fever	MEFV	AR	Turkish	5	99	401
Fanconi anemia, complementation group A	FANCA	AR	General Population	345	99	34401
Fanconi anemia, complementation group A	FANCA	AR	Sephardic Jewish - Moroccan, Tunisian	133	99	13201
Fanconi anemia, complementation group A	FANCA	AR	Spanish Roma	64	99	6301
Fanconi anemia, complementation group C	FANCC	AR	General Population	<500	99	<49901
Fanconi anemia, complementation group C	FANCC	AR	Ashkenazi Jewish	98	99	9701
Fanconi anemia, complementation group G	FANCG	AR	General Population	<500	99	<49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Fanconi anemia, complementation group G	FANCG	AR	African American	100	99	9901
Fragile X syndrome	FMR1	XL	Asian	<500	99	<49901
Fragile X syndrome	FMR1	XL	Hispanic	<500	99	<49901
Fragile X syndrome	FMR1	XL	African American	251	99	25001
Fragile X syndrome	FMR1	XL	General Population	250	99	24901
Fragile X syndrome	FMR1	XL	Caucasian	178	99	17701
Fragile X syndrome	FMR1	XL	Ashkenazi Jewish	58	99	5701
Free sialic acid storage disorders	SLC17A5	AR	General Population	<500	99	<49901
Free sialic acid storage disorders	SLC17A5	AR	Canadian Inuit	129	99	12801
Free sialic acid storage disorders	SLC17A5	AR	Swedish	125	99	12401
Free sialic acid storage disorders	SLC17A5	AR	Finnish	100	99	9901
Fukuyama congenital muscular dystrophy	FKTN	AR	General Population	<500	99	<49901
Fukuyama congenital muscular dystrophy	FKTN	AR	Japanese	188	99	18701
Fukuyama congenital muscular dystrophy	FKTN	AR	Ashkenazi Jewish	150	99	14901
Fumarase deficiency	FH	AR	General Population	<500	99	<49901
Galactosemia	GALT	AR	East Asian	<500	99	<49901
Galactosemia	GALT	AR	Hispanic	305	99	30401
Galactosemia	GALT	AR	Ashkenazi Jewish	172	99	17101
Galactosemia	GALT	AR	General Population	110	99	10901
Galactosemia	GALT	AR	Northern European Caucasian	108	99	10701
Galactosemia	GALT	AR	Caucasian	108	99	10701
Galactosemia	GALT	AR	African American	78	99	7701
Galactosemia	GALT	AR	Irish Travellers	11	99	1001
Galactosemia, type II / Galactokinase deficiency	GALK1	AR	Asian	<500	99	<49901
Galactosemia, type II / Galactokinase deficiency	GALK1	AR	General Population	122	99	12101
Galactosemia, type II / Galactokinase deficiency	GALK1	AR	Roma	47	99	4601
Gaucher disease	GBA	AR	Caucasian	164	95	3261
Gaucher disease	GBA	AR	General Population	153	95	3041
Gaucher disease	GBA	AR	Ashkenazi Jewish	18	95	341
Gitelman syndrome	SLC12A3	AR	General Population	100	99	9901
GLB1-related disorders	GLB1	AR	Caucasian	278	99	27701
GLB1-related disorders	GLB1	AR	General Population	158	99	15701
GLB1-related disorders	GLB1	AR	South Brazil	65	99	6401
GLB1-related disorders	GLB1	AR	Roma	50	99	4901
GLB1-related disorders	GLB1	AR	Maltese	30	99	2901
Glucose-6-phosphate dehydrogenase deficiency	G6PD	XL	General Population	30	99	2901
Glucose-6-phosphate dehydrogenase deficiency	G6PD	XL	African American	5	99	401
Glutaric acidemia, type I	GCDH	AR	Caucasian	172	99	17101
Glutaric acidemia, type I	GCDH	AR	General Population	112	99	11101
Glutaric acidemia, type I	GCDH	AR	African American	36	99	3501
Glutaric acidemia, type I	GCDH	AR	Lumbee Native Americans	16	99	1501
Glutaric acidemia, type I	GCDH	AR	Lancaster County Amish	9	99	801
Glutaric acidemia, type I	GCDH	AR	Oji-Cree First Nations (N. Manitoba)	8	99	701
Glycine encephalopathy / Nonketotic hyperglycinemia	GLDC	AR	Caucasian	140	99	13901
Glycine encephalopathy / Nonketotic hyperglycinemia	GLDC	AR	General Population	135	99	13401
Glycine encephalopathy, AMT-related	AMT	AR	Caucasian	271	99	27001
Glycine encephalopathy, AMT-related	AMT	AR	General Population	262	99	26101
Glycogen storage disease, type Ia	G6PC1	AR	Asian	192	99	19101
Glycogen storage disease, type Ia	G6PC1	AR	General Population	177	99	17601
Glycogen storage disease, type Ia	G6PC1	AR	Caucasian	177	99	17601
Glycogen storage disease, type Ia	G6PC1	AR	Ashkenazi Jewish	71	99	7001

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Glycogen storage disease, type Ia	G6PC1	AR	Ashkenazi Jewish	71	99	7001
Glycogen storage disease, type Ib / IIw	SLC37A4	AR	Caucasian	<500	99	<49901
Glycogen storage disease, type Ib / IIw	SLC37A4	AR	General Population	354	99	35301
Glycogen storage disease, type II / Pompe disease	GAA	AR	General Population	132	99	13101
Glycogen storage disease, type II / Pompe disease	GAA	AR	Asian	112	99	11101
Glycogen storage disease, type II / Pompe disease	GAA	AR	Caucasian	100	99	9901
Glycogen storage disease, type II / Pompe disease	GAA	AR	African American	70	99	6901
Glycogen storage disease, type II / Pompe disease	GAA	AR	Ashkenazi Jewish	58	99	5701
Glycogen storage disease, type III	AGL	AR	General Population	159	99	15801
Glycogen storage disease, type III	AGL	AR	Sephardic Jewish - Moroccan	37	99	3601
Glycogen storage disease, type III	AGL	AR	Faroese	28	99	2701
Glycogen storage disease, type IV / Adult polyglucosan body disease	GBE1	AR	General Population	387	99	38601
Glycogen storage disease, type IV / Adult polyglucosan body disease	GBE1	AR	Caucasian	144	99	14301
Glycogen storage disease, type IV / Adult polyglucosan body disease	GBE1	AR	Ashkenazi Jewish	68	99	6701
Glycogen storage disease, type V	PYGM	AR	Caucasian	191	99	19001
Glycogen storage disease, type V	PYGM	AR	General Population	191	99	19001
Glycogen storage disease, type V	PYGM	AR	Sephardic Jewish - Kurdish	84	99	8301
Glycogen storage disease, type VII	PFKM	AR	General Population	<500	99	<49901
Glycogen storage disease, type VII	PFKM	AR	Ashkenazi Jewish	250	99	24901
GNE myopathy	GNE	AR	Ashkenazi Jewish	<500	99	<49901
GNE myopathy	GNE	AR	Caucasian	<500	99	<49901
GNE myopathy	GNE	AR	General Population	<500	99	<49901
GNE myopathy	GNE	AR	Asian	58	99	5701
GNE myopathy	GNE	AR	Sephardic Jewish - Iranian, Syrian	12	99	1101
GRACILE syndrome	BCS1L	AR	Caucasian	407	99	40601
GRACILE syndrome	BCS1L	AR	General Population	111	99	11001
GRACILE syndrome	BCS1L	AR	Finnish	108	99	10701
Guanidinoacetate methyltransferase deficiency	GAMT	AR	General Population	<500	99	<49901
Guanidinoacetate methyltransferase deficiency	GAMT	AR	Portuguese	125	99	12401
Hereditary fructose intolerance	ALDOB	AR	Hispanic	<500	99	<49901
Hereditary fructose intolerance	ALDOB	AR	African American	406	99	40501
Hereditary fructose intolerance	ALDOB	AR	Caucasian	80	99	7901
Hereditary fructose intolerance	ALDOB	AR	General Population	55	99	5401
Hereditary hemochromatosis, type 2	HJV	AR	Caucasian	<500	99	<49901
Hereditary hemochromatosis, type 2	HJV	AR	General Population	<500	99	<49901
Hereditary hemochromatosis, type 3	TFR2	AR	General Population	<500	99	<49901
Hermansky-Pudlak syndrome, type 1	HPS1	AR	General Population	<500	99	<49901
Hermansky-Pudlak syndrome, type 1	HPS1	AR	Puerto Rican	59	99	5801
Hermansky-Pudlak syndrome, type 3	HPS3	AR	General Population	<500	99	<49901
Hermansky-Pudlak syndrome, type 3	HPS3	AR	Ashkenazi Jewish	235	99	23401
Holocarboxylase synthetase deficiency	HLCS	AR	Caucasian	<500	99	<49901
Holocarboxylase synthetase deficiency	HLCS	AR	General Population	<500	99	<49901
Holocarboxylase synthetase deficiency	HLCS	AR	Asian	158	99	15701
Holocarboxylase synthetase deficiency	HLCS	AR	Faroese	20	99	1901
Homocystinuria caused by methylenetetrahydrofolate reductase (MTHFR) deficiency	MTHFR	AR	General Population	158	99	15701
Homocystinuria caused by methylenetetrahydrofolate reductase (MTHFR) deficiency	MTHFR	AR	Sephardic Jewish - Bukharian	39	99	3801
Homocystinuria, CBS-related	CBS	AR	General Population	224	99	22301
Homocystinuria, CBS-related	CBS	AR	Caucasian	52	99	5101
Homocystinuria, CBS-related	CBS	AR	Qatari	21	99	2001
Homocystinuria, type cblE	MTRR	AR	General Population	<500	99	<49901
Homocystinuria, type cblE	MTRR	AR	Caucasian	<500	99	<49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Hydrolethalus syndrome	HYLS1	AR	General Population	455	99	45401
Hydrolethalus syndrome	HYLS1	AR	Finnish	50	99	4901
Hypohidrotic ectodermal dysplasia, X-linked	EDA	XL	General Population	3800	99	379901
Hypophosphatasia	ALPL	AR	Northern European Caucasian	274	99	27301
Hypophosphatasia	ALPL	AR	East Asian	203	99	20201
Hypophosphatasia	ALPL	AR	Asian	203	99	20201
Hypophosphatasia	ALPL	AR	General Population	158	99	15701
Hypophosphatasia	ALPL	AR	Mennonite	25	99	2401
Infantile cerebral and cerebellar atrophy	MED17	AR	General Population	<500	99	<49901
Infantile cerebral and cerebellar atrophy	MED17	AR	Sephardic Jewish - Bukharian, Kurdish	20	99	1901
Isovaleric acidemia	IVD	AR	General Population	250	99	24901
Isovaleric acidemia	IVD	AR	Caucasian	144	99	14301
Isovaleric acidemia	IVD	AR	Asian	75	99	7401
Joubert syndrome 2	TMEM216	AR	General Population	<500	99	<49901
Joubert syndrome 2	TMEM216	AR	Ashkenazi Jewish	110	99	10901
Junctional epidermolysis bullosa, LAMA3-related	LAMA3	AR	General Population	<500	99	<49901
Junctional epidermolysis bullosa, LAMB3-related	LAMB3	AR	General Population	407	99	40601
Junctional epidermolysis bullosa, LAMC2-related	LAMC2	AR	General Population	<500	99	<49901
Juvenile retinoschisis, X-linked	RS1	XL	General Population	2500	99	249901
Krabbe disease	GALC	AR	Asian	<500	99	<49901
Krabbe disease	GALC	AR	General Population	150	99	14901
Krabbe disease	GALC	AR	Druze Northern Israel	6	99	501
Krabbe disease	GALC	AR	Muslim Arab (Jerusalem)	6	99	501
LAMA2 muscular dystrophy	LAMA2	AR	General Population	87	99	8601
Leber congenital amaurosis 13	RDH12	AR	General Population	456	99	45501
Leber congenital amaurosis 2	RPE65	AR	General Population	228	99	22701
Leber congenital amaurosis 2	RPE65	AR	Sephardic Jewish - North African	90	99	8901
Leber congenital amaurosis 5	LCA5	AR	General Population	<500	99	<49901
Leber congenital amaurosis, CEP290-related / CEP290-related conditions	CEP290	AR	General Population	185	99	18401
Lethal congenital contracture syndrome 1	GLE1	AR	General Population	<500	99	<49901
Lethal congenital contracture syndrome 1	GLE1	AR	Finnish	100	99	9901
Leukoencephalopathy with vanishing white matter 5	EIF2B5	AR	General Population	<500	99	<49901
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	General Population	<500	99	<49901
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	Hispanic	260	99	25901
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	East Asian	232	99	23101
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	Northern European Caucasian	103	99	10201
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	Caucasian	103	99	10201
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	Amish	50	99	4901
Limb-girdle muscular dystrophy, type 2B	DYSF	AR	General Population	311	99	31001
Limb-girdle muscular dystrophy, type 2B	DYSF	AR	Caucasian	158	99	15701
Limb-girdle muscular dystrophy, type 2B	DYSF	AR	Sephardic Jewish, Libyan, Moroccan, Tunisian, Bulgarian	14	99	1301
Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	FKRP	AR	General Population	158	99	15701
Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	FKRP	AR	Norwegian	116	99	11501
Limb-girdle muscular dystrophy, type 3	SGCA	AR	General Population	<500	99	<49901
Limb-girdle muscular dystrophy, type 3	SGCA	AR	Caucasian	290	99	28901
Limb-girdle muscular dystrophy, type 3	SGCA	AR	Northern European Caucasian	160	99	15901
Limb-girdle muscular dystrophy, type 3	SGCA	AR	Finnish	150	99	14901
Limb-girdle muscular dystrophy, type 4	SGCB	AR	Northern European Caucasian	<500	99	<49901
Limb-girdle muscular dystrophy, type 4	SGCB	AR	General Population	<500	99	<49901
Limb-girdle muscular dystrophy, type 4	SGCB	AR	Caucasian	406	99	40501

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Limb-girdle muscular dystrophy, type 5	SGCG	AR	Northern European Caucasian	<500	99	<49901
Limb-girdle muscular dystrophy, type 5	SGCG	AR	General Population	354	99	35301
Limb-girdle muscular dystrophy, type 5	SGCG	AR	Moroccan	250	99	24901
Limb-girdle muscular dystrophy, type 5	SGCG	AR	Roma	96	99	9501
Lipoid congenital adrenal hyperplasia	STAR	AR	General Population	<500	99	<49901
Lipoid congenital adrenal hyperplasia	STAR	AR	East Asian	177	99	17601
Lipoprotein lipase deficiency	LPL	AR	General Population	<500	99	<49901
Lipoprotein lipase deficiency	LPL	AR	Caucasian	<500	99	<49901
Lipoprotein lipase deficiency	LPL	AR	Asian	189	99	18801
Lipoprotein lipase deficiency	LPL	AR	French Canadian - Other	139	99	13801
Lipoprotein lipase deficiency	LPL	AR	French Canadian - Saguenay Lac-St. Jean	46	99	4501
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA	AR	Caucasian	254	99	25301
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA	AR	Finnish	240	99	23901
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA	AR	General Population	138	99	13701
Lysinuric protein intolerance	SLC7A7	AR	General Population	<500	99	<49901
Lysinuric protein intolerance	SLC7A7	AR	Finnish	122	99	12101
Lysinuric protein intolerance	SLC7A7	AR	Japanese	119	99	11801
Lysosomal acid lipase deficiency	LIPA	AR	General Population	<500	99	<49901
Lysosomal acid lipase deficiency	LIPA	AR	Ashkenazi Jewish	<500	99	<49901
Lysosomal acid lipase deficiency	LIPA	AR	Caucasian	145	99	14401
Lysosomal acid lipase deficiency	LIPA	AR	Sephardic Jewish - Iranian	26	99	2501
Maple syrup urine disease, type 1A	BCKDHA	AR	General Population	321	99	32001
Maple syrup urine disease, type 1A	BCKDHA	AR	Caucasian	320	99	31901
Maple syrup urine disease, type 1A	BCKDHA	AR	Portuguese Roma	71	99	7001
Maple syrup urine disease, type 1A	BCKDHA	AR	Mennonite	10	99	901
Maple syrup urine disease, type 1B	BCKDHB	AR	Caucasian	433	99	43201
Maple syrup urine disease, type 1B	BCKDHB	AR	General Population	364	99	36301
Maple syrup urine disease, type 1B	BCKDHB	AR	Asian	163	99	16201
Maple syrup urine disease, type 1B	BCKDHB	AR	Ashkenazi Jewish	97	99	9601
Maple syrup urine disease, type 2	DBT	AR	General Population	321	99	32001
Medium chain acyl-CoA dehydrogenase deficiency	ACADM	AR	Asian	178	99	17701
Medium chain acyl-CoA dehydrogenase deficiency	ACADM	AR	Caucasian	64	99	6301
Medium chain acyl-CoA dehydrogenase deficiency	ACADM	AR	General Population	35	99	3401
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	AR	General Population	<500	99	<49901
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	AR	Libyan Jewish	40	99	3901
Menkes disease	ATP7A	XL	General Population	26000	99	2599901
Metachromatic leukodystrophy due to saposin B deficiency	PSAP	AR	General Population	<500	99	<49901
Metachromatic leukodystrophy, ARSA-related	ARSA	AR	Ashkenazi Jewish	<500	99	<49901
Metachromatic leukodystrophy, ARSA-related	ARSA	AR	General Population	100	99	9901
Metachromatic leukodystrophy, ARSA-related	ARSA	AR	Sephardic Jewish - Yemenite	46	99	4501
Metachromatic leukodystrophy, ARSA-related	ARSA	AR	Navajo	25	99	2401
Methylmalonic aciduria, MMAA-related	MMAA	AR	General Population	316	99	31501
Methylmalonic aciduria, MMAA-related	MMAA	AR	Caucasian	316	99	31501
Methylmalonic aciduria, MMAB-related	MMAB	AR	Caucasian	456	99	45501
Methylmalonic aciduria, MMAB-related	MMAB	AR	General Population	456	99	45501
Methylmalonic aciduria, MMUT-related	MMUT	AR	General Population	383	99	38201
Methylmalonic aciduria, MMUT-related	MMUT	AR	Hispanic	383	99	38201
Methylmalonic aciduria, MMUT-related	MMUT	AR	Caucasian	224	99	22301
Methylmalonic aciduria, MMUT-related	MMUT	AR	African American	177	99	17601
Methylmalonic aciduria, MMUT-related	MMUT	AR	Asian	53	99	5201
Microphthalmia / Anophthalmia	VSX2	AR	General Population	<500	99	<49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Microphthalmia / Anophthalmia	VSX2	AR	Sephardic Jewish - Iranian, Syrian	145	99	14401
Mitochondrial complex I deficiency, ACAD9-related	ACAD9	AR	General Population	<500	99	<49901
Mitochondrial complex I deficiency, nuclear type 16	NDUFAF5	AR	General Population	<500	99	<49901
Mitochondrial complex I deficiency, nuclear type 16	NDUFAF5	AR	Ashkenazi Jewish	290	99	28901
Mitochondrial complex I deficiency, nuclear type 9	NDUFS6	AR	General Population	<500	99	<49901
Mitochondrial complex I deficiency, nuclear type 9	NDUFS6	AR	Caucasus Jewish	24	99	2301
Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type	LRPPRC	AR	General Population	<500	99	<49901
Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type	LRPPRC	AR	French Canadian - Saguenay Lac-St. Jean	23	99	2201
Mitochondrial DNA depletion syndrome 1, MNGIE type	TYMP	AR	Caucasian	<500	99	<49901
Mitochondrial DNA depletion syndrome 1, MNGIE type	TYMP	AR	General Population	<500	99	<49901
Mitochondrial DNA depletion syndrome 1, MNGIE type	TYMP	AR	Sephardic Jewish - Iranian	158	99	15701
Mitochondrial trifunctional protein deficiency, HADHB-related	HADHB	AR	General Population	146	99	14501
MKS1-related disorders	MKS1	AR	General Population	260	99	25901
MKS1-related disorders	MKS1	AR	Caucasian	260	99	25901
MKS1-related disorders	MKS1	AR	Finnish	47	99	4601
MPV17-related mitochondrial DNA (mtDNA) maintenance defect	MPV17	AR	General Population	<500	99	<49901
MPV17-related mitochondrial DNA (mtDNA) maintenance defect	MPV17	AR	Navajo	20	99	1901
Mucolipidosis II and mucolipidosis III alpha/beta	GNPTAB	AR	Asian	389	99	38801
Mucolipidosis II and mucolipidosis III alpha/beta	GNPTAB	AR	Caucasian	225	99	22401
Mucolipidosis II and mucolipidosis III alpha/beta	GNPTAB	AR	General Population	158	99	15701
Mucolipidosis III gamma	GNPTG	AR	General Population	<500	99	<49901
Mucolipidosis III gamma	GNPTG	AR	Caucasian	273	99	27201
Mucolipidosis IV	MCOLN1	AR	General Population	<500	99	<49901
Mucolipidosis IV	MCOLN1	AR	Ashkenazi Jewish	89	99	8801
Mucopolysaccharidosis, type I / Hurler syndrome	IDUA	AR	General Population	158	99	15701
Mucopolysaccharidosis, type I / Hurler syndrome	IDUA	AR	Northern European Caucasian	145	99	14401
Mucopolysaccharidosis, type II / Hunter syndrome	IDS	XL	General Population	60000	90	599991
Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A	SGSH	AR	General Population	415	99	41401
Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A	SGSH	AR	Caucasian	253	99	25201
Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	NAGLU	AR	General Population	<500	99	<49901
Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	NAGLU	AR	Caucasian	346	99	34501
Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	NAGLU	AR	Asian	298	99	29701
Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	HGSNAT	AR	Asian	<500	99	<49901
Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	HGSNAT	AR	General Population	482	99	48101
Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	HGSNAT	AR	Caucasian	259	99	25801
Mucopolysaccharidosis, type IIID / Sanfilippo syndrome D	GNS	AR	General Population	<500	99	<49901
Mucopolysaccharidosis, type IX / Hyaluronidase deficiency	HYAL1	AR	General Population	<500	99	<49901
Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	ARSB	AR	Asian	423	99	42201
Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	ARSB	AR	General Population	291	99	29001
Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	ARSB	AR	Caucasian	273	99	27201
Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIA	ETFA	AR	General Population	<500	99	<49901
Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIC	ETFDH	AR	General Population	250	99	24901
Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIC	ETFDH	AR	Asian	87	99	8601
Multiple sulfatase deficiency	SUMF1	AR	General Population	<500	99	<49901
Multiple sulfatase deficiency	SUMF1	AR	Ashkenazi Jewish	279	99	27801
Muscular dystrophy-dystroglycanopathy, type A, 3	POMGNT1	AR	General Population	462	99	46101
Muscular dystrophy-dystroglycanopathy, type A, 3	POMGNT1	AR	Finnish	111	99	11001
Myopathy, lactic acidosis, and sideroblastic anemia	PUS1	AR	Sephardic Jewish - Iranian	<500	99	<49901
Myopathy, lactic acidosis, and sideroblastic anemia	PUS1	AR	General Population	<500	99	<49901
N-acetylglutamate synthase deficiency	NAGS	AR	General Population	<500	99	<49901
Nemaline myopathy 2	NEB	AR	General Population	224	95	4461

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Nemaline myopathy 2	NEB	AR	Ashkenazi Jewish	168	95	3341
Nemaline myopathy 2	NEB	AR	Finnish	112	95	2221
Nephrogenic diabetes insipidus	AQP2	AR	General Population	<500	99	<49901
Neuronal ceroid lipofuscinosis, CLN3-related	CLN3	AR	Caucasian	188	99	18701
Neuronal ceroid lipofuscinosis, CLN3-related	CLN3	AR	General Population	145	99	14401
Neuronal ceroid lipofuscinosis, CLN5-related	CLN5	AR	General Population	317	99	31601
Neuronal ceroid lipofuscinosis, CLN5-related	CLN5	AR	Finnish	289	99	28801
Neuronal ceroid lipofuscinosis, CLN6-related	CLN6	AR	General Population	261	99	26001
Neuronal ceroid lipofuscinosis, CLN8-related	CLN8	AR	General Population	349	99	34801
Neuronal ceroid lipofuscinosis, CLN8-related	CLN8	AR	Finnish	135	99	13401
Neuronal ceroid lipofuscinosis, MFSD8-related	MFSD8	AR	General Population	<500	99	<49901
Neuronal ceroid lipofuscinosis, PPT1-related	PPT1	AR	General Population	368	99	36701
Neuronal ceroid lipofuscinosis, PPT1-related	PPT1	AR	Finnish	70	99	6901
Neuronal ceroid lipofuscinosis, TPP1-related	TPP1	AR	General Population	314	99	31301
Neuronal ceroid lipofuscinosis, TPP1-related	TPP1	AR	Newfoundland	59	99	5801
Niemann-Pick disease, type C1	NPC1	AR	Asian	404	99	40301
Niemann-Pick disease, type C1	NPC1	AR	General Population	282	99	28101
Niemann-Pick disease, type C1	NPC1	AR	Caucasian	185	99	18401
Niemann-Pick disease, type C2	NPC2	AR	General Population	<500	99	<49901
Niemann-Pick disease, types A/B	SMPD1	AR	Caucasian	244	99	24301
Niemann-Pick disease, types A/B	SMPD1	AR	General Population	196	99	19501
Niemann-Pick disease, types A/B	SMPD1	AR	Ashkenazi Jewish	115	99	11401
Nijmegen breakage syndrome	NBN	AR	General Population	<500	99	<49901
Nijmegen breakage syndrome	NBN	AR	Caucasian	155	99	15401
Nonsyndromic hearing loss and deafness (DFNB) 1	GJB2	AR	General Population	42	99	4101
Nonsyndromic hearing loss and deafness (DFNB) 1	GJB2	AR	Caucasian	30	99	2901
Nonsyndromic hearing loss and deafness (DFNB) 1	GJB2	AR	Ashkenazi Jewish	21	99	2001
Nonsyndromic hearing loss and deafness (DFNB) 1	GJB2	AR	East Asian	10	99	901
Nonsyndromic hearing loss and deafness (DFNB) 77	LOXHD1	AR	General Population	<500	99	<49901
Nonsyndromic hearing loss and deafness (DFNB) 77	LOXHD1	AR	Ashkenazi Jewish	180	99	17901
Odonto-onycho-dermal dysplasia / Schopf-Schulz-Passarge syndrome	WNT10A	AR	General Population	305	99	30401
Omenn syndrome	DCLRE1C	AR	Northern European Caucasian	<500	99	<49901
Omenn syndrome	DCLRE1C	AR	General Population	<500	99	<49901
Omenn syndrome	DCLRE1C	AR	Navajo and Apache Native Americans	10	99	901
Ornithine aminotransferase deficiency	OAT	AR	General Population	<500	99	<49901
Ornithine aminotransferase deficiency	OAT	AR	Sephardic Jewish - Iraqi, Syrian	177	99	17601
Ornithine aminotransferase deficiency	OAT	AR	Finnish	147	99	14601
Ornithine transcarbamylase deficiency, X-linked	OTC	XL	General Population	30000	99	2999901
Ornithine translocase deficiency	SLC25A15	AR	General Population	<500	99	<49901
Ornithine translocase deficiency	SLC25A15	AR	French Canadian	20	99	1901
Ornithine translocase deficiency	SLC25A15	AR	Metis from Saskatchewan	19	99	1801
Osteopetrosis, infantile malignant, TCIRG1-related	TCIRG1	AR	Ashkenazi Jewish	350	99	34901
Osteopetrosis, infantile malignant, TCIRG1-related	TCIRG1	AR	General Population	316	99	31501
Osteopetrosis, infantile malignant, TCIRG1-related	TCIRG1	AR	Costa Rican	86	99	8501
Osteopetrosis, infantile malignant, TCIRG1-related	TCIRG1	AR	Chuvashiya	60	99	5901
Pendred syndrome	SLC26A4	AR	Caucasian	88	99	8701
Pendred syndrome	SLC26A4	AR	General Population	80	99	7901
Pendred syndrome	SLC26A4	AR	African American	76	99	7501
Pendred syndrome	SLC26A4	AR	Asian	74	99	7301
Pendred syndrome	SLC26A4	AR	Northern European Caucasian	60	99	5901
Peroxisomal acyl-CoA oxidase deficiency	ACOX1	AR	General Population	<500	99	<49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Phenylalanine hydroxylase deficiency	PAH	AR	Finnish	225	99	22401
Phenylalanine hydroxylase deficiency	PAH	AR	Ashkenazi Jewish	225	99	22401
Phenylalanine hydroxylase deficiency	PAH	AR	Hispanic American	163	99	16201
Phenylalanine hydroxylase deficiency	PAH	AR	African American	143	99	14201
Phenylalanine hydroxylase deficiency	PAH	AR	Asian	78	99	7701
Phenylalanine hydroxylase deficiency	PAH	AR	General Population	65	99	6401
Phenylalanine hydroxylase deficiency	PAH	AR	Caucasian	50	99	4901
Phenylalanine hydroxylase deficiency	PAH	AR	Southern European Caucasian	40	99	3901
Phenylalanine hydroxylase deficiency	PAH	AR	Irish	34	99	3301
Phenylalanine hydroxylase deficiency	PAH	AR	Turkish	32	99	3101
Phenylalanine hydroxylase deficiency	PAH	AR	Sicilian	26	99	2501
Phenylalanine hydroxylase deficiency	PAH	AR	Sephardic Jewish - Iranian, Bukharian, Kavkazi, Tunisian, Moroccan	18	99	1701
Phosphoglycerate dehydrogenase deficiency	PHGDH	AR	General Population	<500	99	<49901
Phosphoglycerate dehydrogenase deficiency	PHGDH	AR	Ashkenazi Jewish	453	99	45201
Pontocerebellar hypoplasia, type 1A	VRK1	AR	General Population	<500	99	<49901
Pontocerebellar hypoplasia, type 1A	VRK1	AR	Ashkenazi Jewish	225	99	22401
Pontocerebellar hypoplasia, type 6	RARS2	AR	General Population	<500	99	<49901
Pontocerebellar hypoplasia, type 6	RARS2	AR	Sephardic Jewish - Iraqi, Syrian, Tunisian	<500	99	<49901
Primary ciliary dyskinesia, DNAH5-related	DNAH5	AR	Ashkenazi Jewish	174	99	17301
Primary ciliary dyskinesia, DNAH5-related	DNAH5	AR	General Population	120	99	11901
Primary ciliary dyskinesia, DNAI1-related	DNAI1	AR	Ashkenazi Jewish	352	99	35101
Primary ciliary dyskinesia, DNAI1-related	DNAI1	AR	General Population	182	99	18101
Primary ciliary dyskinesia, DNAI2-related	DNAI2	AR	General Population	<500	99	<49901
Primary ciliary dyskinesia, DNAI2-related	DNAI2	AR	Ashkenazi Jewish	200	99	19901
Primary hyperoxaluria, type I	AGXT	AR	General Population	158	99	15701
Primary hyperoxaluria, type II	GRHPR	AR	General Population	<500	99	<49901
Primary hyperoxaluria, type III	HOGA1	AR	General Population	309	99	30801
Progressive cerebello-cerebral atrophy	SEPSECS	AR	General Population	<500	99	<49901
Progressive cerebello-cerebral atrophy	SEPSECS	AR	Sephardic Jewish - Moroccan, Iraqi	41	99	4001
Progressive familial intrahepatic cholestasis 2	ABCB11	AR	General Population	158	99	15701
Propionic acidemia, PCCA-related	PCCA	AR	Caucasian	380	99	37901
Propionic acidemia, PCCA-related	PCCA	AR	General Population	224	99	22301
Propionic acidemia, PCCA-related	PCCA	AR	Asian	162	99	16101
Propionic acidemia, PCCB-related	PCCB	AR	General Population	224	99	22301
Propionic acidemia, PCCB-related	PCCB	AR	Caucasian	202	99	20101
Propionic acidemia, PCCB-related	PCCB	AR	Asian	145	99	14401
PRPS1-related disorders	PRPS1	XL	General Population	<500000	99	49999901
Pycnodysostosis	CTSK	AR	General Population	439	99	43801
Pyruvate carboxylase deficiency	PC	AR	General Population	250	99	24901
Pyruvate dehydrogenase E1-alpha deficiency	PDHA1	XL	General Population	<750000	99	<74999901
Pyruvate dehydrogenase E1-beta deficiency	PDHB	AR	General Population	<500	99	<49901
Renal tubular acidosis and deafness, ATP6V1B1-related	ATP6V1B1	AR	General Population	<500	99	<49901
Renal tubular acidosis and deafness, ATP6V1B1-related	ATP6V1B1	AR	Sephardic Jewish - Syrian	140	99	13901
Retinitis pigmentosa 25	EYS	AR	Ashkenazi Jewish	<500	99	<49901
Retinitis pigmentosa 25	EYS	AR	General Population	129	99	12801
Retinitis pigmentosa 25	EYS	AR	Caucasian	53	99	5201
Retinitis pigmentosa 25	EYS	AR	Sephardic Jewish - Moroccan	42	99	4101
Retinitis pigmentosa 26	CERKL	AR	General Population	137	99	13601
Retinitis pigmentosa 26	CERKL	AR	Sephardic Jewish - Yemenite	24	99	2301
Retinitis pigmentosa 28	FAM161A	AR	General Population	289	99	28801

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Retinitis pigmentosa 28	FAM161A	AR	Ashkenazi Jewish	214	99	21301
Retinitis pigmentosa 28	FAM161A	AR	Sephardic Jewish, Libyan, Moroccan, Tunisian, Bulgarian	41	99	4001
Retinitis pigmentosa 59	DHDDS	AR	General Population	<500	99	<49901
Retinitis pigmentosa 59	DHDDS	AR	Ashkenazi Jewish	117	99	11601
Rhizomelic chondrodysplasia punctata, type 1	PEX7	AR	General Population	<500	99	<49901
Rhizomelic chondrodysplasia punctata, type 1	PEX7	AR	Caucasian	158	99	15701
Rhizomelic chondrodysplasia punctata, type 3	AGPS	AR	General Population	<500	99	<49901
Roberts-SC phocomelia syndrome	ESCO2	AR	General Population	<500	99	<49901
Sandhoff disease	HEXB	AR	Ashkenazi Jewish	<500	99	<49901
Sandhoff disease	HEXB	AR	General Population	278	99	27701
Sandhoff disease	HEXB	AR	Caucasian	235	99	23401
Sandhoff disease	HEXB	AR	Argentinian Creole	64	99	6301
Schimke immunoosseous dysplasia	SMARCAL1	AR	General Population	<500	99	<49901
Severe combined immunodeficiency, RAG2-related	RAG2	AR	General Population	<500	99	<49901
Severe combined immunodeficiency, RAG2-related	RAG2	AR	Sephardic Jewish - Iraqi	<500	99	<49901
Severe combined immunodeficiency, X-linked	IL2RG	XL	General Population	69000	99	6899901
Severe congenital neutropenia 5	VPS45	AR	General Population	<500	99	<49901
Sjogren-Larsson syndrome	ALDH3A2	AR	Northern European Caucasian	223	99	22201
Sjogren-Larsson syndrome	ALDH3A2	AR	General Population	223	99	22201
Sjogren-Larsson syndrome	ALDH3A2	AR	Swedish	204	99	20301
Skeletal dysplasia, SLC26A2-related	SLC26A2	AR	General Population	158	99	15701
Skeletal dysplasia, SLC26A2-related	SLC26A2	AR	Finnish	50	99	4901
Smith-Lemli-Opitz syndrome	DHCR7	AR	Asian	<500	99	<49901
Smith-Lemli-Opitz syndrome	DHCR7	AR	General Population	100	99	9901
Smith-Lemli-Opitz syndrome	DHCR7	AR	African American	93	99	9201
Smith-Lemli-Opitz syndrome	DHCR7	AR	Northern European Caucasian	50	99	4901
Smith-Lemli-Opitz syndrome	DHCR7	AR	Caucasian	50	99	4901
Smith-Lemli-Opitz syndrome	DHCR7	AR	Ashkenazi Jewish	36	99	3501
Smith-Lemli-Opitz syndrome	DHCR7	AR	Ashkenazi Jewish	36	99	3501
Spastic ataxia, Charlevoix-Saguenay type	SACS	AR	General Population	<500	99	<49901
Spastic ataxia, Charlevoix-Saguenay type	SACS	AR	Caucasian	450	99	44901
Spastic ataxia, Charlevoix-Saguenay type	SACS	AR	French Canadian - Charlevoix-Saguenay	21	99	2001
Spastic paraplegia, type 15	ZFYVE26	AR	General Population	<500	99	<49901
Spinal muscular atrophy	SMN1	AR	Hispanic	117	99	11601
Spinal muscular atrophy	SMN1	AR	African American	72	99	7101
Spinal muscular atrophy	SMN1	AR	Hispanic American	68	99	6701
Spinal muscular atrophy	SMN1	AR	Ashkenazi Jewish	67	99	6601
Spinal muscular atrophy	SMN1	AR	East Asian	59	99	5801
Spinal muscular atrophy	SMN1	AR	South Asian	59	99	5801
Spinal muscular atrophy	SMN1	AR	Southeast Asian	59	99	5801
Spinal muscular atrophy	SMN1	AR	Asian	59	99	5801
Spinal muscular atrophy	SMN1	AR	General Population	54	99	5301
Spinal muscular atrophy	SMN1	AR	Northern European Caucasian	47	99	4601
Spinal muscular atrophy	SMN1	AR	Caucasian	47	99	4601
Spondylothoracic dysostosis and spondylocostal dysostosis 2	MESP2	AR	General Population	224	99	22301
Spondylothoracic dysostosis and spondylocostal dysostosis 2	MESP2	AR	Puerto Rican	55	99	5401
Steel syndrome	COL27A1	AR	General Population	<500	99	<49901
Steroid resistant nephrotic syndrome, type 1	NPHS1	AR	General Population	325	99	32401
Steroid resistant nephrotic syndrome, type 1	NPHS1	AR	Finnish	45	99	4401
Steroid resistant nephrotic syndrome, type 1	NPHS1	AR	Groffdale Conference Mennonites	12	99	1101

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Steroid-resistant nephrotic syndrome, type 2	NPHS2	AR	General Population	377	99	37601
Stuve-Wiedemann syndrome	LIFR	AR	General Population	<500	99	<49901
Tay-Sachs disease	HEXA	AR	African American	271	99	27001
Tay-Sachs disease	HEXA	AR	General Population	250	99	24901
Tay-Sachs disease	HEXA	AR	Caucasian	182	99	18101
Tay-Sachs disease	HEXA	AR	Asian	126	99	12501
Tay-Sachs disease	HEXA	AR	Sephardic Jewish - Moroccan, Iraqi	125	99	12401
Tay-Sachs disease	HEXA	AR	French Canadian - Other	73	99	7201
Tay-Sachs disease	HEXA	AR	Irish	41	99	4001
Tay-Sachs disease	HEXA	AR	Ashkenazi Jewish	27	99	2601
Tay-Sachs disease	HEXA	AR	French Canadian	13	99	1201
Tay-Sachs disease	HEXA	AR	French Canadian - Gaspesie	13	99	1201
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	TECPR2	AR	General Population	<500	99	<49901
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	TECPR2	AR	Sephardic Jewish - Bukharian	27	99	2601
Tyrosine hydroxylase deficiency	TH	AR	General Population	<500	99	<49901
Tyrosine hydroxylase deficiency	TH	AR	Asian	416	99	41501
Tyrosine hydroxylase deficiency	TH	AR	Caucasian	224	99	22301
Tyrosinemia, type I	FAH	AR	Asian	<500	99	<49901
Tyrosinemia, type I	FAH	AR	African American	478	99	47701
Tyrosinemia, type I	FAH	AR	Caucasian	333	99	33201
Tyrosinemia, type I	FAH	AR	Ashkenazi Jewish	150	99	14901
Tyrosinemia, type I	FAH	AR	General Population	100	99	9901
Tyrosinemia, type I	FAH	AR	French Canadian (Qubec	66	99	6501
Tyrosinemia, type I	FAH	AR	French Canadian - Saguenay Lac-St. Jean	25	99	2401
Tyrosinemia, type II	TAT	AR	General Population	<500	99	<49901
Usher syndrome, type 1B	MYO7A	AR	African American	<500	99	<49901
Usher syndrome, type 1B	MYO7A	AR	General Population	206	99	20501
Usher syndrome, type 1B	MYO7A	AR	Caucasian	145	99	14401
Usher syndrome, type 1B	MYO7A	AR	Japanese	123	99	12201
Usher syndrome, type 1B	MYO7A	AR	Asian	62	99	6101
Usher syndrome, type 1C	USH1C	AR	General Population	353	99	35201
Usher syndrome, type 1C	USH1C	AR	French Canadian / Acadian	227	99	22601
Usher syndrome, type 1C	USH1C	AR	Acadian	41	99	4001
Usher syndrome, type 1D	CDH23	AR	General Population	202	99	20101
Usher syndrome, type 1F	PCDH15	AR	General Population	395	99	39401
Usher syndrome, type 1F	PCDH15	AR	Ashkenazi Jewish	78	99	7701
Usher syndrome, type 2A	USH2A	AR	French Canadian	207	99	20601
Usher syndrome, type 2A	USH2A	AR	General Population	126	99	12501
Usher syndrome, type 2A	USH2A	AR	East Asian	113	99	11201
Usher syndrome, type 2A	USH2A	AR	Northern European Caucasian	113	99	11201
Usher syndrome, type 2A	USH2A	AR	Caucasian	73	99	7201
Usher syndrome, type 2A	USH2A	AR	Sephardic Jewish - Iraqi, Iranian	36	99	3501
Usher syndrome, type 3A	CLRN1	AR	General Population	<500	99	<49901
Usher syndrome, type 3A	CLRN1	AR	Ashkenazi Jewish	120	99	11901
Usher syndrome, type 3A	CLRN1	AR	Finnish	70	99	6901
Very long-chain acyl-CoA dehydrogenase deficiency	ACADVL	AR	Asian	194	99	19301
Very long-chain acyl-CoA dehydrogenase deficiency	ACADVL	AR	Caucasian	88	99	8701
Very long-chain acyl-CoA dehydrogenase deficiency	ACADVL	AR	General Population	86	99	8501
Wilson disease	ATP7B	AR	Southern European Caucasian	250	99	24901
Wilson disease	ATP7B	AR	Northern European Caucasian	90	99	8901
Wilson disease	ATP7B	AR	General Population	90	99	8901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency (one in ***)	Detection Rate	Residual Risk (one in ***)
Wilson disease	ATP7B	AR	Caucasian	90	99	8901
Wilson disease	ATP7B	AR	Ashkenazi Jewish	70	99	6901
Wilson disease	ATP7B	AR	Sephardic Jewish - North African, Iraqi, Yemenite, Iranian, Bukharian	65	99	6401
Wilson disease	ATP7B	AR	Sardinian	42	99	4101
Wilson disease	ATP7B	AR	East Asian	27	99	2601
Wilson disease	ATP7B	AR	Canary Islands	25	99	2401
Xeroderma pigmentosum, group A	XPA	AR	General Population	<500	99	<49901
Xeroderma pigmentosum, group C	XPC	AR	General Population	<500	99	<49901
X-linked myotubular myopathy	MTM1	XL	General Population	38000	99	3799901
Zellweger spectrum disorders, PEX10-related	PEX10	AR	Asian	<500	99	<49901
Zellweger spectrum disorders, PEX10-related	PEX10	AR	General Population	<500	99	<49901
Zellweger spectrum disorders, PEX12-related	PEX12	AR	General Population	406	99	40501
Zellweger spectrum disorders, PEX1-related	PEX1	AR	General Population	<500	99	<49901
Zellweger spectrum disorders, PEX1-related	PEX1	AR	Caucasian	147	99	14601
Zellweger spectrum disorders, PEX2-related	PEX2	AR	General Population	<500	99	<49901
Zellweger spectrum disorders, PEX2-related	PEX2	AR	Caucasian	<500	99	<49901
Zellweger spectrum disorders, PEX2-related	PEX2	AR	Ashkenazi Jewish	227	99	22601
Zellweger spectrum disorders, PEX6-related	PEX6	AR	General Population	280	99	27901
Zellweger spectrum disorders, PEX6-related	PEX6	AR	French Canadian	55	99	5401
Zellweger spectrum disorders, PEX6-related	PEX6	AR	Sephardic Jewish - Yemenite	18	99	1701