GeneAware™ (version 4) Residual Risk Table

BAYLOR GENETICS

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
17-beta-hydroxysteroid dehydrogenase deficiency, type III	HSD17B3	AR	General Population	<1 in 500	99%	<1 in 49901
3-beta-hydroxysteroid dehydrogenase deficiency, type II	HSD3B2	AR	General Population	<1 in 500	99%	<1 in 49901
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL	AR	General Population	<1 in 500	99%	<1 in 49901
3-hydroxyacyl-CoA dehydrogenase deficiency	HADH	AR	General Population	<1 in 500	99%	<1 in 49901
3-methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1	AR	General Population	1 in 147	99%	1 in 14601
3-methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1	AR	Caucasian	1 in 137	99%	1 in 13601
3-methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2	AR	General Population	1 in 120	99%	1 in 11901
3-methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2	AR	Caucasian	1 in 112	99%	1 in 11101
3-methylglutaconic aciduria, type III / Costeff syndrome	OPA3	AR	General Population	<1 in 500	99%	<1 in 49901
3-methylglutaconic aciduria, type III / Costeff syndrome	OPA3	AR	Sephardic Jewish - Iraqi	1 in 13	99%	1 in 1201
6-pyruvoyl-tetrahydropterin synthase deficiency	PTS	AR	General Population	<1 in 500	99%	<1 in 49901
6-pyruvoyl-tetrahydropterin synthase deficiency	PTS	AR	East Asian	1 in 158	99%	1 in 15701
6-pyruvoyl-tetrahydropterin synthase deficiency	PTS	AR	Asian	1 in 122	99%	1 in 12101
Abetalipoproteinemia	MTTP	AR	Caucasian	<1 in 500	99%	<1 in 49901
Abetalipoproteinemia	MTTP	AR	General Population	<1 in 500	99%	<1 in 49901
Abetalipoproteinemia	MTTP	AR	Ashkenazi Jewish	1 in 186	99%	1 in 18501
Achromatopsia, CNGB3-related	CNGB3	AR	General Population	1 in 98	99%	1 in 9701
Achromatopsia, CNGB3-related	CNGB3	AR	Caucasian	1 in 91	99%	1 in 9001
Acrodermatitis enteropathica	SLC39A4	AR	General Population	1 in 354	99%	1 in 35301
Action myoclonus renal failure syndrome	SCARB2	AR	General Population	<1 in 500	99%	<1 in 49901
Acute infantile liver failure	TRMU	AR	General Population	<1 in 500	99%	<1 in 49901
Acute infantile liver failure	TRMU	AR	Sephardic Jewish - Yemenite	1 in 34	99%	1 in 3301
Adenosine deaminase deficiency	ADA	AR	General Population	1 in 224	99%	1 in 22301
Adrenoleukodystrophy, X-linked	ABCD1	XL	General Population	1 in 10500	99%	1 in 1049901
Adrenoleukodystrophy, X-linked	ABCD1	XL	Sephardic Jewish	1 in 10500	99%	1 in 1049901
Agenesis of the corpus callosum with peripheral neuropathy	SLC12A6	AR	General Population	<1 in 500	99%	<1 in 49901
Agenesis of the corpus callosum with peripheral neuropathy	SLC12A6	AR	French Canadian	1 in 23	99%	1 in 2201
Aicardi-Goutieres syndrome 2	RNASEH2B	AR	General Population	<1 in 500	99%	<1 in 49901
Aicardi-Goutieres syndrome 3	RNASEH2C	AR	General Population	<1 in 500	99%	<1 in 49901
Aicardi-Goutieres syndrome 4	RNASEH2A	AR	General Population	<1 in 500	99%	<1 in 49901
Aicardi-Goutieres syndrome 5	SAMHD1	AR	General Population	<1 in 500	99%	<1 in 49901
Alpha-1 antitrypsin deficiency	SERPINA1	AR	General Population	1 in 38	99%	1 in 3701
Alpha-1 antitrypsin deficiency	SERPINA1	AR	Ashkenazi Jewish	1 in 24	99%	1 in 2301
Alpha-1 antitrypsin deficiency	SERPINA1	AR	Northern European Caucasian	1 in 15	99%	1 in 1401
Alpha-mannosidosis	MAN2B1	AR	General Population	<1 in 500	99%	<1 in 49901
Alpha-mannosidosis	MAN2B1	AR	Caucasian	1 in 485	99%	1 in 48401
Alpha-mannosidosis	MAN2B1	AR	Northern European Caucasian	1 in 354	99%	1 in 35301
Alpha-thalassemia	HBA1/HBA2	AR	Caucasian	<1 in 500	95%	<1 in 9981
Alpha-thalassemia	HBA1/HBA2	AR	African American	1 in 30	95%	1 in 581
Alpha-thalassemia	HBA1/HBA2	AR	General Population	1 in 25	95%	1 in 481
Alpha-thalassemia	HBA1/HBA2	AR	Asian	1 in 20	95%	1 in 381
Alpha-thalassemia	HBA1/HBA2	AR	East Asian	1 in 16	95%	1 in 301
Alpha-thalassemia	HBA1/HBA2	AR	Southeast Asian	1 in 7	95%	1 in 121

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Alpha-thalassemia	HBA1/HBA2	AR	South Asian	1 in 2	95%	1 in 21
Alpha-thalassemia intellectual disability syndrome, X-linked	ATRX	XL	General Population	<1 in 750000	99%	<1 in 74999901
Alport syndrome, COL4A3-related	COL4A3	AR	General Population	1 in 323	99%	1 in 32201
Alport syndrome, COL4A3-related	COL4A3	AR	Caucasian	1 in 284	99%	1 in 28301
Alport syndrome, COL4A3-related	COL4A3	AR	Ashkenazi Jewish	1 in 189	99%	1 in 18801
Alport syndrome, COL4A4-related	COL4A4	AR	General Population	1 in 353	99%	1 in 35201
Alport syndrome, COL4A5-related, X-linked	COL4A5	XL	General Population	1 in 47000	99%	1 in 4699901
Alstrom syndrome	ALMS1	AR	General Population	<1 in 500	99%	<1 in 49901
Amish infantile epilepsy syndrome	ST3GAL5	AR	General Population	<1 in 500	99%	<1 in 49901
Argininemia	ARG1	AR	General Population	<1 in 500	99%	<1 in 49901
Argininosuccinic aciduria	ASL	AR	Finnish	1 in 190	99%	1 in 18901
Argininosuccinic aciduria	ASL	AR	General Population	1 in 133	99%	1 in 13201
Aromatase deficiency	CYP19A1	AR	General Population	<1 in 500	99%	<1 in 49901
Arthrogryposis, mental retardation, and seizures	SLC35A3	AR	General Population	<1 in 500	99%	<1 in 49901
Arthrogryposis, mental retardation, and seizures	SLC35A3	AR	Ashkenazi Jewish	1 in 453	99%	1 in 45201
Asparagine synthetase deficiency	ASNS	AR	General Population	<1 in 500	99%	<1 in 49901
Asparagine synthetase deficiency	ASNS	AR	Sephardic Jewish - Iranian	1 in 80	99%	1 in 7901
Aspartylglycosaminuria	AGA	AR	General Population	<1 in 500	99%	<1 in 49901
Aspartylglycosaminuria	AGA	AR	Caucasian	<1 in 500	99%	<1 in 49901
Aspartylglycosaminuria	AGA	AR	Finnish	1 in 36	99%	1 in 3501
Ataxia with isolated vitamin E deficiency	TTPA	AR	General Population	<1 in 500	99%	<1 in 49901
Ataxia with isolated vitamin E deficiency	TTPA	AR	Caucasian	<1 in 500	99%	<1 in 49901
Ataxia-telangiectasia	ATM	AR	Ashkenazi Jewish	<1 in 500	99%	<1 in 49901
Ataxia-telangiectasia	ATM	AR	General Population	1 in 100	99%	1 in 9901
Ataxia-telangiectasia	ATM	AR	Sephardic Jewish - Moroccan	1 in 81	99%	1 in 8001
Ataxia-telangiectasia-like disorder 1	MRE11	AR	General Population	<1 in 500	99%	<1 in 49901
Atransferrinemia	TF	AR	General Population	1 in 116	99%	1 in 11501
Autoimmune polyglandular syndrome, type 1	AIRE	AR	General Population	1 in 354	99%	1 in 35301
Autoimmune polyglandular syndrome, type 1	AIRE	AR	Finnish	1 in 79	99%	1 in 7801
Autoimmune polyglandular syndrome, type 1	AIRE	AR	Sardinian	1 in 60	99%	1 in 5901
Autoimmune polyglandular syndrome, type 1	AIRE	AR	Sephardic Jewish - Iranian	1 in 27	99%	1 in 2601
Autosomal recessive congenital ichthyosis	TGM1	AR	General Population	1 in 301	99%	1 in 30001
Autosomal recessive congenital ichthyosis	TGM1	AR	Caucasian	1 in 253	99%	1 in 25201
Autosomal recessive congenital ichthyosis	TGM1	AR	Norwegian	1 in 151	99%	1 in 15001
Autosomal recessive polycystic kidney disease	PKHD1	AR	General Population	1 in 144	99%	1 in 14301
Autosomal recessive polycystic kidney disease	PKHD1	AR	Ashkenazi Jewish	1 in 106	99%	1 in 10501
Autosomal recessive polycystic kidney disease	PKHD1	AR	Caucasian	1 in 100	99%	1 in 9901
Autosomal recessive polycystic kidney disease	PKHD1	AR	South African Afrikaner	1 in 52	99%	1 in 5101
Autosomal recessive primary microcephaly 1	MCPH1	AR	Northern European Caucasian	<1 in 500	99%	<1 in 49901
Autosomal recessive primary microcephaly 1	MCPH1	AR	General Population	1 in 146	99%	1 in 14501
Autosomal recessive spinocerebellar ataxia, type 10	ANO10	AR	General Population	1 in 93	99%	1 in 9201
Bardet-Biedl syndrome 1	BBS1	AR	General Population	1 in 265	99%	1 in 26401
Bardet-Biedl syndrome 1	BBS1	AR	Faroese	1 in 30	99%	1 in 2901
Bardet-Biedl syndrome 10	BBS10	AR	General Population	1 in 447	99%	1 in 44601
Bardet-Biedl syndrome 12	BBS12	AR	General Population	<1 in 500	99%	<1 in 49901
Bardet-Biedl syndrome 2	BBS2	AR	General Population	<1 in 500	99%	<1 in 49901
Bardet-Biedl syndrome 2	BBS2	AR	Ashkenazi Jewish	1 in 135	99%	1 in 13401
Bardet-Biedl syndrome 2	BBS2	AR	Hutterites	1 in 22	99%	1 in 2101
Bardet-Biedl syndrome 4	BBS4	AR	General Population	<1 in 500	99%	<1 in 49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Bardet-Biedl syndrome 6	MKKS	AR	General Population	1 in 219	99%	1 in 21801
Bardet-Biedl syndrome 7	BBS7	AR	General Population	<1 in 500	99%	<1 in 49901
Bardet-Biedl syndrome 8	TTC8	AR	General Population	<1 in 500	99%	<1 in 49901
Bardet-Biedl syndrome 9	BBS9	AR	General Population	<1 in 500	99%	<1 in 49901
Bare lymphocyte syndrome, type II	CIITA	AR	General Population	<1 in 500	99%	<1 in 49901
Barth syndrome	TAFAZZIN	XL	General Population	1 in 225000	99%	1 in 22499901
Bartter syndrome, type 4A	BSND	AR	General Population	<1 in 500	99%	<1 in 49901
Bernard-Soulier syndrome, type A	GP1BA	AR	General Population	<1 in 500	99%	<1 in 49901
Bernard-Soulier syndrome, type C	GP9	AR	General Population	<1 in 500	99%	<1 in 49901
Beta hemoglobinopathies	HBB	AR	Caucasian	1 in 373	99%	1 in 37201
Beta hemoglobinopathies	HBB	AR	General Population	1 in 129	99%	1 in 12801
Beta hemoglobinopathies	HBB	AR	Hispanic	1 in 83	99%	1 in 8201
Beta hemoglobinopathies	HBB	AR	East Asian	1 in 78	99%	1 in 7701
Beta hemoglobinopathies	HBB	AR	Southern European Caucasian	1 in 59	99%	1 in 5801
Beta hemoglobinopathies	HBB	AR	Asian	1 in 54	99%	1 in 5301
Beta hemoglobinopathies	HBB	AR	South Asian	1 in 32	99%	1 in 3101
Beta hemoglobinopathies	HBB	AR	Southeast Asian	1 in 30	99%	1 in 2901
Beta hemoglobinopathies	HBB	AR	Mediterranean	1 in 28	99%	1 in 2701
Beta hemoglobinopathies	HBB	AR	African American	1 in 10	99%	1 in 901
Beta hemoglobinopathies	HBB	AR	Middle Eastern	1 in 5	99%	1 in 401
Beta-ketothiolase deficiency	ACAT1	AR	Caucasian	1 in 354	99%	1 in 35301
Beta-ketothiolase deficiency	ACAT1	AR	General Population	1 in 347	99%	1 in 34601
Beta-ketothiolase deficiency	ACAT1	AR	Asian	1 in 289	99%	1 in 28801
Beta-ureidopropionase deficiency	UPB1	AR	General Population	<1 in 500	99%	<1 in 49901
Bilateral frontoparietal polymicrogyria	ADGRG1	AR	General Population	<1 in 500	99%	<1 in 49901
Biotinidase deficiency	BTD	AR	General Population	1 in 120	99%	1 in 11901
Biotinidase deficiency	BTD	AR	Hispanic	1 in 30	99%	1 in 2901
Biotinidase deficiency	BTD	AR	Caucasian	1 in 12	99%	1 in 1101
Biotin-thiamine-responsive basal ganglia disease	SLC19A3	AR	Middle Eastern	<1 in 500	99%	<1 in 49901
Biotin-thiamine-responsive basal ganglia disease	SLC19A3	AR	General Population	1 in 109	99%	1 in 10801
Bloom syndrome	BLM	AR	General Population	<1 in 500	99%	<1 in 49901
Bloom syndrome	BLM	AR	Ashkenazi Jewish	1 in 140	99%	1 in 13901
Canavan disease	ASPA	AR	General Population	<1 in 500	99%	<1 in 49901
Canavan disease	ASPA	AR	Ashkenazi Jewish	1 in 60	99%	1 in 5901
Carbamoyl phosphate synthetase I deficiency	CPS1	AR	General Population	<1 in 500	99%	<1 in 49901
Carbamoyl phosphate synthetase I deficiency	CPS1	AR	Asian	1 in 447	99%	1 in 44601
Carbamoyl phosphate synthetase I deficiency	CPS1	AR	Caucasian	1 in 284	99%	1 in 28301
Carnitine deficiency, systemic primary	SLC22A5	AR	General Population	1 in 200	99%	1 in 19901
Carnitine deficiency, systemic primary	SLC22A5	AR	Caucasian	1 in 110	99%	1 in 10901
Carnitine deficiency, systemic primary	SLC22A5	AR	East Asian	1 in 100	99%	1 in 9901
Carnitine deficiency, systemic primary	SLC22A5	AR	Asian	1 in 100	99%	1 in 9901
Carnitine deficiency, systemic primary	SLC22A5	AR	Faroese	1 in 20	99%	1 in 1901
Carnitine palmitoyltransferase I deficiency	CPT1A	AR	General Population	<1 in 500	99%	<1 in 49901
Carnitine palmitoyltransferase I deficiency	CPT1A	AR	Hutterites	1 in 16	99%	1 in 1501
Carnitine palmitoyltransferase II deficiency	CPT2	AR	General Population	<1 in 500	99%	<1 in 49901
Carnitine palmitoyltransferase II deficiency	CPT2	AR	Asian	<1 in 500	99%	<1 in 49901
Carnitine palmitoyltransferase II deficiency	CPT2	AR	African American	1 in 308	99%	1 in 30701
Carnitine palmitoyltransferase II deficiency	CPT2	AR	Caucasian	1 in 200	99%	1 in 19901
Carnitine palmitoyltransferase II deficiency	CPT2	AR	Ashkenazi Jewish	1 in 51	99%	1 in 5001

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Carnitine-acylcarnitine translocase deficiency	SLC25A20	AR	General Population	<1 in 500	99%	<1 in 49901
Carpenter syndrome	RAB23	AR	General Population	<1 in 500	99%	<1 in 49901
Carpenter syndrome	RAB23	AR	Caucasian	<1 in 500	99%	<1 in 49901
Cartilage-hair hypoplasia	RMRP	AR	General Population	<1 in 500	99%	<1 in 49901
Cartilage-hair hypoplasia	RMRP	AR	Finnish	1 in 76	99%	1 in 7501
Cartilage-hair hypoplasia	RMRP	AR	Amish	1 in 19	99%	1 in 1801
Cerebrooculofacioskeletal syndrome 1 / Cockayne syndrome, type B	ERCC6	AR	General Population	<1 in 500	99%	<1 in 49901
Cerebrotendinous xanthomatosis	CYP27A1	AR	Southern European Caucasian	<1 in 500	99%	<1 in 49901
Cerebrotendinous xanthomatosis	CYP27A1	AR	General Population	1 in 115	99%	1 in 11401
Cerebrotendinous xanthomatosis	CYP27A1	AR	Sephardic Jewish - Moroccan	1 in 5	99%	1 in 401
Charcot-Marie-Tooth disease, type 1X	GJB1	XL	General Population	1 in 7000	99%	1 in 699901
Charcot-Marie-Tooth disease, type 4D	NDRG1	AR	General Population	<1 in 500	99%	<1 in 49901
Charcot-Marie-Tooth disease, type 4D	NDRG1	AR	Roma	1 in 22	99%	1 in 2101
Chediak-Higashi syndrome	LYST	AR	General Population	<1 in 500	99%	<1 in 49901
Choreoacanthocytosis	VPS13A	AR	General Population	<1 in 500	99%	<1 in 49901
Choreoacanthocytosis	VPS13A	AR	Ashkenazi Jewish	<1 in 500	99%	<1 in 49901
Choroideremia, X-linked	CHM	XL	General Population	1 in 25000	99%	1 in 2499901
Chronic granulomatous disease 4	CYBA	AR	General Population	<1 in 500	99%	<1 in 49901
Chronic granulomatous disease 4	CYBA	AR	Sephardic Jewish - Moroccan	1 in 13	99%	1 in 1201
Chronic granulomatous disease, X-linked	CYBB	XL	General Population	1 in 150000	99%	1 in 14999901
Ciliopathies, RPGRIP1L-related	RPGRIP1L	AR	General Population	1 in 259	99%	1 in 25801
Citrin deficiency / Citrullinemia, type II	SLC25A13	AR	Caucasian	<1 in 500	99%	<1 in 49901
Citrin deficiency / Citrullinemia, type II	SLC25A13	AR	General Population	<1 in 500	99%	<1 in 49901
Citrin deficiency / Citrullinemia, type II	SLC25A13	AR	Asian	1 in 123	99%	1 in 12201
Citrin deficiency / Citrullinemia, type II	SLC25A13	AR	East Asian	1 in 65	99%	1 in 6401
Citrullinemia, type I	ASS1	AR	Caucasian	1 in 195	99%	1 in 19401
Citrullinemia, type I	ASS1	AR	Asian	1 in 123	99%	1 in 12201
Citrullinemia, type I	ASS1	AR	General Population	1 in 119	99%	1 in 11801
Cockayne syndrome, type A	ERCC8	AR	General Population	<1 in 500	99%	<1 in 49901
Cohen syndrome	VPS13B	AR	General Population	<1 in 500	99%	<1 in 49901
Combined malonic and methylmalonic aciduria	ACSF3	AR	General Population	1 in 86	99%	1 in 8501
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	MMACHC	AR	General Population	1 in 138	99%	1 in 13701
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	MMACHC	AR	Caucasian	1 in 138	99%	1 in 13701
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	MMACHC	AR	Asian	1 in 113	99%	1 in 11201
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	MMACHC	AR	East Asian	1 in 112	99%	1 in 11101
Combined methylmalonic aciduria and homocystinuria, cblD type / Cobalamin D deficiency	MMADHC	AR	Caucasian	<1 in 500	99%	<1 in 49901
Combined methylmalonic aciduria and homocystinuria, cblD type / Cobalamin D deficiency	MMADHC	AR	General Population	<1 in 500	99%	<1 in 49901
Combined oxidative phosphorylation deficiency 1	GFM1	AR	General Population	<1 in 500	99%	<1 in 49901
Combined oxidative phosphorylation deficiency 3	TSFM	AR	General Population	<1 in 500	99%	<1 in 49901
Combined oxidative phosphorylation deficiency 3	TSFM	AR	Finnish	1 in 80	99%	1 in 7901
Combined oxidative phosphorylation deficiency 6	AIFM1	AR	General Population	<1 in 500	99%	<1 in 49901
Combined pituitary hormone deficiency, type 2	PROP1	AR	General Population	1 in 141	99%	1 in 14001
Combined pituitary hormone deficiency, type 3	LHX3	AR	General Population	<1 in 500	99%	<1 in 49901
Congenital adrenal hyperplasia (CAH) due to 11-beta-hydroxylase deficiency	CYP11B1	AR	General Population	1 in 158	99%	1 in 15701
Congenital adrenal hyperplasia (CAH) due to 17-alpha-hydroxylase deficiency	CYP17A1	AR	General Population	<1 in 500	99%	<1 in 49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	CYP21A2	AR	Caucasian	1 in 67	98%	1 in 3301
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	CYP21A2	AR	General Population	1 in 61	98%	1 in 3001
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	CYP21A2	AR	Ashkenazi Jewish	1 in 40	98%	1 in 1951
Congenital adrenal insufficiency, CYP11A1-related	CYP11A1	AR	General Population	1 in 114	99%	1 in 11301
Congenital amegakaryocytic thrombocytopenia	MPL	AR	General Population	1 in 415	99%	1 in 41401
Congenital amegakaryocytic thrombocytopenia	MPL	AR	Caucasian	1 in 266	99%	1 in 26501
Congenital amegakaryocytic thrombocytopenia	MPL	AR	Ashkenazi Jewish	1 in 57	99%	1 in 5601
Congenital disorder of glycosylation, type Ia	PMM2	AR	Asian	1 in 449	99%	1 in 44801
Congenital disorder of glycosylation, type Ia	PMM2	AR	General Population	1 in 124	99%	1 in 12301
Congenital disorder of glycosylation, type Ia	PMM2	AR	Ashkenazi Jewish	1 in 61	99%	1 in 6001
Congenital disorder of glycosylation, type Ia	PMM2	AR	Northern European Caucasian	1 in 60	99%	1 in 5901
Congenital disorder of glycosylation, type Ia	PMM2	AR	Caucasian	1 in 42	99%	1 in 4101
Congenital disorder of glycosylation, type Ib	MPI	AR	General Population	<1 in 500	99%	<1 in 49901
Congenital disorder of glycosylation, type Ic	ALG6	AR	General Population	<1 in 500	99%	<1 in 49901
Congenital hydrocephalus 1	CCDC88C	AR	General Population	1 in 137	99%	1 in 13601
Congenital ichthyosis, ABCA12-related	ABCA12	AR	General Population	<1 in 500	99%	<1 in 49901
Congenital insensitivity to pain with anhidrosis	NTRK1	AR	Sephardic Jewish - Moroccan	<1 in 500	99%	<1 in 49901
Congenital insensitivity to pain with anhidrosis	NTRK1	AR	General Population	<1 in 500	99%	<1 in 49901
Congenital insensitivity to pain with anhidrosis	NTRK1	AR	Asian	1 in 387	99%	1 in 38601
Congenital myasthenic syndrome, CHAT-related	CHAT	AR	General Population	<1 in 500	99%	<1 in 49901
Congenital myasthenic syndrome, CHRNE-related	CHRNE	AR	General Population	<1 in 500	99%	<1 in 49901
Congenital myasthenic syndrome, CHRNE-related	CHRNE	AR	Caucasian	1 in 383	99%	1 in 38201
Congenital myasthenic syndrome, COLQ-related	COLQ	AR	General Population	1 in 430	99%	1 in 42901
Congenital myasthenic syndrome, DOK7-related	DOK7	AR	General Population	1 in 454	99%	1 in 45301
Congenital myasthenic syndrome, DOK7-related	DOK7	AR	French Canadian	1 in 353	99%	1 in 35201
Congenital myasthenic syndrome, DOK7-related	DOK7	AR	Northern European Caucasian	1 in 290	99%	1 in 28901
Congenital myasthenic syndrome, DOK7-related	DOK7	AR	Caucasian	1 in 290	99%	1 in 28901
Congenital myasthenic syndrome, RAPSN-related	RAPSN	AR	Sephardic Jewish - Iraqi, Iranian	<1 in 500	99%	<1 in 49901
Congenital myasthenic syndrome, RAPSN-related	RAPSN	AR	General Population	1 in 252	99%	1 in 25101
Congenital myasthenic syndrome, RAPSN-related	RAPSN	AR	Caucasian	1 in 176	99%	1 in 17501
Congenital neutropenia, HAX1-related	HAX1	AR	General Population	<1 in 500	99%	<1 in 49901
Corneal dystrophy and perceptive deafness syndrome	SLC4A11	AR	General Population	<1 in 500	99%	<1 in 49901
Corticosterone methyloxidase deficiency	CYP11B2	AR	General Population	<1 in 500	99%	<1 in 49901
Corticosterone methyloxidase deficiency	CYP11B2	AR	Sephardic Jewish - Iranian	1 in 30	99%	1 in 2901
CRB1-related retinal dystrophies	CRB1	AR	General Population	1 in 112	99%	1 in 11101
Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome	SLC6A8	XL	General Population	1 in 20600	99%	1 in 2059901
Cystic fibrosis	CFTR	AR	South Asian	1 in 90	99%	1 in 8901
Cystic fibrosis	CFTR	AR	Southeast Asian	1 in 90	99%	1 in 8901
Cystic fibrosis	CFTR	AR	East Asian	1 in 90	99%	1 in 8901
Cystic fibrosis	CFTR	AR	African American	1 in 61	99%	1 in 6001
Cystic fibrosis	CFTR	AR	Hispanic	1 in 46	99%	1 in 4501
Cystic fibrosis	CFTR	AR	Northern European Caucasian	1 in 25	99%	1 in 2401
Cystic fibrosis	CFTR	AR	Ashkenazi Jewish	1 in 25	99%	1 in 2401
Cystic fibrosis	CFTR	AR	Caucasian	1 in 25	99%	1 in 2401
Cystic fibrosis	CFTR	AR	General Population	1 in 25	99%	1 in 2401
Cystinosis	CTNS	AR	African American	<1 in 500	99%	<1 in 49901
Cystinosis	CTNS	AR	Asian	<1 in 500	99%	<1 in 49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Cystinosis	CTNS	AR	Hispanic	<1 in 500	99%	<1 in 49901
Cystinosis	CTNS	AR	Caucasian	1 in 220	99%	1 in 21901
Cystinosis	CTNS	AR	General Population	1 in 158	99%	1 in 15701
Cystinosis	CTNS	AR	Sephardic Jewish - Moroccan	1 in 100	99%	1 in 9901
Cystinosis	CTNS	AR	French Canadian - Saguenay Lac-St. Jean	1 in 39	99%	1 in 3801
D-bifunctional protein deficiency	HSD17B4	AR	General Population	1 in 158	99%	1 in 15701
DCX-related disorders	DCX	XL	General Population	<1 in 750000	99%	<1 in 74999901
Dent disease	CLCN5	AR	General Population	<1 in 500	99%	<1 in 49901
Desbuquois dysplasia, type I	CANT1	AR	General Population	<1 in 500	99%	<1 in 49901
Dihydrolipoamide dehydrogenase deficiency	DLD	AR	General Population	<1 in 500	99%	<1 in 49901
Dihydrolipoamide dehydrogenase deficiency	DLD	AR	Ashkenazi Jewish	1 in 94	99%	1 in 9301
Dihydropyrimidine dehydrogenase deficiency	DPYD	AR	East Asian	1 in 50	99%	1 in 4901
Dihydropyrimidine dehydrogenase deficiency	DPYD	AR	General Population	1 in 20	99%	1 in 1901
Donnai-Barrow syndrome	LRP2	AR	General Population	1 in 213	99%	1 in 21201
Duchenne/Becker muscular dystrophy, X-linked	DMD	XL	General Population	1 in 4200	99%	1 in 419901
Dyskeratosis congenita, RTEL1-related	RTEL1	AR	General Population	<1 in 500	99%	<1 in 49901
Dyskeratosis congenita, RTEL1-related	RTEL1	AR	Ashkenazi Jewish	1 in 165	99%	1 in 16401
Dyskeratosis congenita, X-linked	DKC1	XL	General Population	<1 in 750000	99%	<1 in 74999901
Dystrophic epidermolysis bullosa, COL7A1-related	COL7A1	AR	General Population	1 in 370	99%	1 in 36901
Ehlers-Danlos syndrome, dermatosparaxis type	ADAMTS2	AR	General Population	<1 in 500	99%	<1 in 49901
Ehlers-Danlos syndrome, dermatosparaxis type	ADAMTS2	AR	Ashkenazi Jewish	1 in 248	99%	1 in 24701
Ellis-van Creveld syndrome	EVC	AR	General Population	1 in 345	99%	1 in 34401
Ellis-van Creveld syndrome	EVC	AR	Lancaster County Amish	1 in 12	99%	1 in 1101
Ellis-van Creveld syndrome	EVC2	AR	General Population	1 in 122	99%	1 in 12101
Emery-Dreifuss muscular dystrophy, X-linked	EMD	XL	General Population	1 in 250000	99%	1 in 24999901
Enhanced S-cone syndrome	NR2E3	AR	General Population	1 in 204	99%	1 in 20301
Enhanced S-cone syndrome	NR2E3	AR	Ashkenazi Jewish	1 in 100	99%	1 in 9901
ERCC2-related conditions	ERCC2	AR	General Population	<1 in 500	99%	<1 in 49901
Ethylmalonic encephalopathy	ETHE1	AR	General Population	<1 in 500	99%	<1 in 49901
Fabry disease, X-linked	GLA	XL	General Population	1 in 1500	99%	1 in 149901
Factor IX deficiency / Hemophilia B	F9	XL	General Population	1 in 10000	99%	1 in 999901
Factor XI deficiency / Hemophilia C	F11	AR	Asian	1 in 163	99%	1 in 16201
Factor XI deficiency / Hemophilia C	F11	AR	Caucasian	1 in 101	99%	1 in 10001
Factor XI deficiency / Hemophilia C	F11	AR	General Population	1 in 92	99%	1 in 9101
Factor XI deficiency / Hemophilia C	F11	AR	Ashkenazi Jewish	1 in 11	99%	1 in 1001
Factory VIII deficiency / Hemophilia A	F8	XL	General Population	1 in 2000	95%	1 in 39981
Familial dysautonomia	ELP1	AR	General Population	<1 in 500	99%	<1 in 49901
Familial dysautonomia	ELP1	AR	Ashkenazi Jewish	1 in 34	99%	1 in 3301
Familial hemophagocytic lymphohistiocytosis 2	PRF1	AR	General Population	<1 in 500	99%	<1 in 49901
Familial hemophagocytic lymphohistiocytosis 4	STX11	AR	General Population	1 in 354	99%	1 in 35301
Familial hemophagocytic lymphohistiocytosis 5	STXBP2	AR	General Population	1 in 296	99%	1 in 29501
Familial hypercholesterolemia, LDLRAP1-related	LDLRAP1	AR	General Population	<1 in 500	99%	<1 in 49901
Familial hypercholesterolemia, LDLRAP1-related	LDLRAP1	AR	Sardinian	1 in 143	99%	1 in 14201
Familial hypercholesterolemia, LDLR-related	LDLR	AR	General Population	<1 in 500	99%	<1 in 49901
Familial hypercholesterolemia, LDLR-related	LDLR	AR	French Canadian	1 in 267	99%	1 in 26601
Familial hypercholesterolemia, LDLR-related	LDLR	AR	Caucasian	1 in 200	99%	1 in 19901
Familial hypercholesterolemia, LDLR-related	LDLR	AR	Finnish	1 in 143	99%	1 in 14201
Familial hypercholesterolemia, LDLR-related	LDLR	AR	South African Afrikaner	1 in 70	99%	1 in 6901
Familial hypercholesterolemia, LDLR-related	LDLR	AR	Ashkenazi Jewish	1 in 69	99%	1 in 6801

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	General Population	1 in 112	99%	1 in 11101
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	Ashkenazi Jewish	1 in 52	99%	1 in 5101
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	Finnish	1 in 29	99%	1 in 2801
Familial hyperinsulinism, KCNJ11-related	KCNJ11	AR	General Population	<1 in 500	99%	<1 in 49901
Familial Mediterranean fever	MEFV	AR	General Population	1 in 115	99%	1 in 11401
Familial Mediterranean fever	MEFV	AR	Sephardic Jewish	1 in 14	99%	1 in 1301
Familial Mediterranean fever	MEFV	AR	Ashkenazi Jewish	1 in 13	99%	1 in 1201
Familial Mediterranean fever	MEFV	AR	Armenian	1 in 5	99%	1 in 401
Familial Mediterranean fever	MEFV	AR	Turkish	1 in 5	99%	1 in 401
Fanconi anemia, complementation group A	FANCA	AR	General Population	1 in 345	99%	1 in 34401
Fanconi anemia, complementation group A	FANCA	AR	Sephardic Jewish - Moroccan, Tunisian	1 in 133	99%	1 in 13201
Fanconi anemia, complementation group A	FANCA	AR	Spanish Roma	1 in 64	99%	1 in 6301
Fanconi anemia, complementation group B	FANCB	XL	General Population	<1 in 750000	99%	<1 in 74999901
Fanconi anemia, complementation group C	FANCC	AR	General Population	<1 in 500	99%	<1 in 49901
Fanconi anemia, complementation group C	FANCC	AR	Ashkenazi Jewish	1 in 98	99%	1 in 9701
Fanconi anemia, complementation group D2	FANCD2	AR	General Population	<1 in 500	98%	<1 in 24951
Fanconi anemia, complementation group E	FANCE	AR	General Population	<1 in 500	99%	<1 in 49901
Fanconi anemia, complementation group F	FANCF	AR	General Population	<1 in 500	99%	<1 in 49901
Fanconi anemia, complementation group G	FANCG	AR	General Population	<1 in 500	99%	<1 in 49901
Fanconi anemia, complementation group G	FANCG	AR	African American	1 in 100	99%	1 in 9901
Fanconi anemia, complementation group I	FANCI	AR	General Population	<1 in 500	99%	<1 in 49901
Fanconi anemia, complementation group L	FANCL	AR	General Population	<1 in 500	99%	<1 in 49901
Farber lipogranulomatosis	ASAH1	AR	General Population	<1 in 500	99%	<1 in 49901
Fragile X syndrome	FMR1	AR	Asian	<1 in 500	99%	<1 in 49901
Fragile X syndrome	FMR1	AR	Hispanic	<1 in 500	99%	<1 in 49901
Fragile X syndrome	FMR1	AR	African American	1 in 251	99%	1 in 25001
Fragile X syndrome	FMR1	AR	General Population	1 in 250	99%	1 in 24901
Fragile X syndrome	FMR1	AR	Caucasian	1 in 178	99%	1 in 17701
Fragile X syndrome	FMR1	AR	Ashkenazi Jewish	1 in 58	99%	1 in 5701
Fragile XE syndrome	AFF2	XL	General Population	1 in 18750	99%	1 in 1874901
Fraser syndrome, type 3	GRIP1	AR	General Population	1 in 83	99%	1 in 8201
Free sialic acid storage disorders	SLC17A5	AR	General Population	<1 in 500	99%	<1 in 49901
Free sialic acid storage disorders	SLC17A5	AR	Canadian Inuit	1 in 129	99%	1 in 12801
Free sialic acid storage disorders	SLC17A5	AR	Swedish	1 in 125	99%	1 in 12401
Free sialic acid storage disorders	SLC17A5	AR	Finnish	1 in 100	99%	1 in 9901
Friedreich ataxia	FXN	AR	General Population	1 in 250	96%	1 in 6226
Friedreich ataxia	FXN	AR	Caucasian	1 in 80	96%	1 in 1976
Fukuyama congenital muscular dystrophy	FKTN	AR	General Population	<1 in 500	99%	<1 in 49901
Fukuyama congenital muscular dystrophy	FKTN	AR	Japanese	1 in 188	99%	1 in 18701
Fukuyama congenital muscular dystrophy	FKTN	AR	Ashkenazi Jewish	1 in 150	99%	1 in 14901
Fumarase deficiency	FH	AR	General Population	<1 in 500	99%	<1 in 49901
GABA-transaminase deficiency	ABAT	AR	General Population	<1 in 500	99%	<1 in 49901
Galactosemia	GALT	AR	East Asian	<1 in 500	99%	<1 in 49901
Galactosemia	GALT	AR	Hispanic	1 in 305	99%	1 in 30401
Galactosemia	GALT	AR	Ashkenazi Jewish	1 in 172	99%	1 in 17101
Galactosemia	GALT	AR	General Population	1 in 110	99%	1 in 10901
Galactosemia	GALT	AR	Northern European Caucasian	1 in 108	99%	1 in 10701
Galactosemia	GALT	AR	Caucasian	1 in 108	99%	1 in 10701
Galactosemia	GALT	AR	African American	1 in 78	99%	1 in 7701

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Galactosemia	GALT	AR	Irish Travellers	1 in 11	99%	1 in 1001
Galactosemia, type II / Galactokinase deficiency	GALK1	AR	Asian	<1 in 500	99%	<1 in 49901
Galactosemia, type II / Galactokinase deficiency	GALK1	AR	General Population	1 in 122	99%	1 in 12101
Galactosemia, type II / Galactokinase deficiency	GALK1	AR	Roma	1 in 47	99%	1 in 4601
Galactosialidosis	CTSA	AR	General Population	<1 in 500	99%	<1 in 49901
Gaucher disease	GBA	AR	Caucasian	1 in 164	95%	1 in 3261
Gaucher disease	GBA	AR	General Population	1 in 153	95%	1 in 3041
Gaucher disease	GBA	AR	Ashkenazi Jewish	1 in 18	95%	1 in 341
Gitelman syndrome	SLC12A3	AR	General Population	1 in 100	99%	1 in 9901
GLB1-related disorders	GLB1	AR	Caucasian	1 in 278	99%	1 in 27701
GLB1-related disorders	GLB1	AR	General Population	1 in 158	99%	1 in 15701
GLB1-related disorders	GLB1	AR	South Brazil	1 in 65	99%	1 in 6401
GLB1-related disorders	GLB1	AR	Roma	1 in 50	99%	1 in 4901
GLB1-related disorders	GLB1	AR	Maltese	1 in 30	99%	1 in 2901
Glucose-6-phosphate dehydrogenase deficiency	G6PD	AR	General Population	1 in 30	99%	1 in 2901
Glucose-6-phosphate dehydrogenase deficiency	G6PD	AR	African American	1 in 5	99%	1 in 401
Glutaric acidemia, type I	GCDH	AR	Caucasian	1 in 172	99%	1 in 17101
Glutaric acidemia, type I	GCDH	AR	General Population	1 in 112	99%	1 in 11101
Glutaric acidemia, type I	GCDH	AR	African American	1 in 36	99%	1 in 3501
Glutaric acidemia, type I	GCDH	AR	Lumbee Native Americans	1 in 16	99%	1 in 1501
Glutaric acidemia, type I	GCDH	AR	Lancaster County Amish	1 in 9	99%	1 in 801
Glutaric acidemia, type I	GCDH	AR	Oji-Cree First Nations (N. Manitoba)	1 in 8	99%	1 in 701
Glycine encephalopathy / Nonketotic hyperglycinemia	GLDC	AR	Caucasian	1 in 140	99%	1 in 13901
Glycine encephalopathy / Nonketotic hyperglycinemia	GLDC	AR	General Population	1 in 135	99%	1 in 13401
Glycine encephalopathy, AMT-related	AMT	AR	Caucasian	1 in 271	99%	1 in 27001
Glycine encephalopathy, AMT-related	AMT	AR	General Population	1 in 262	99%	1 in 26101
Glycogen storage disease, type Ia	G6PC1	AR	Asian	1 in 192	99%	1 in 19101
Glycogen storage disease, type Ia	G6PC1	AR	General Population	1 in 177	99%	1 in 17601
Glycogen storage disease, type Ia	G6PC1	AR	Caucasian	1 in 177	99%	1 in 17601
Glycogen storage disease, type Ia	G6PC1	AR	Ashkenazi Jewish	1 in 71	99%	1 in 7001
Glycogen storage disease, type Ia	G6PC1	AR	Ashkenazi Jewish	1 in 71	99%	1 in 7001
Glycogen storage disease, type Ib / IIw	SLC37A4	AR	Caucasian	<1 in 500	99%	<1 in 49901
Glycogen storage disease, type Ib / IIw	SLC37A4	AR	General Population	1 in 354	99%	1 in 35301
Glycogen storage disease, type II / Pompe disease	GAA	AR	General Population	1 in 132	99%	1 in 13101
Glycogen storage disease, type II / Pompe disease	GAA	AR	Asian	1 in 112	99%	1 in 11101
Glycogen storage disease, type II / Pompe disease	GAA	AR	Caucasian	1 in 100	99%	1 in 9901
Glycogen storage disease, type II / Pompe disease	GAA	AR	African American	1 in 70	99%	1 in 6901
Glycogen storage disease, type II / Pompe disease	GAA	AR	Ashkenazi Jewish	1 in 58	99%	1 in 5701
Glycogen storage disease, type III	AGL	AR	General Population	1 in 159	99%	1 in 15801
Glycogen storage disease, type III	AGL	AR	Sephardic Jewish - Moroccan	1 in 37	99%	1 in 3601
Glycogen storage disease, type III	AGL	AR	Faroese	1 in 28	99%	1 in 2701
Glycogen storage disease, type IV / Adult polyglucosan body disease	GBE1	AR	General Population	1 in 387	99%	1 in 38601
Glycogen storage disease, type IV / Adult polyglucosan body disease	GBE1	AR	Caucasian	1 in 144	99%	1 in 14301
Glycogen storage disease, type IV / Adult polyglucosan body disease	GBE1	AR	Ashkenazi Jewish	1 in 68	99%	1 in 6701
Glycogen storage disease, type V	PYGM	AR	Caucasian	1 in 191	99%	1 in 19001
Glycogen storage disease, type V	PYGM	AR	General Population	1 in 191	99%	1 in 19001
Glycogen storage disease, type V	PYGM	AR	Sephardic Jewish - Kurdish	1 in 84	99%	1 in 8301

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Glycogen storage disease, type VII	PFKM	AR	General Population	<1 in 500	99%	<1 in 49901
Glycogen storage disease, type VII	PFKM	AR	Ashkenazi Jewish	1 in 250	99%	1 in 24901
GNE myopathy	GNE	AR	Ashkenazi Jewish	<1 in 500	99%	<1 in 49901
GNE myopathy	GNE	AR	Caucasian	<1 in 500	99%	<1 in 49901
GNE myopathy	GNE	AR	General Population	<1 in 500	99%	<1 in 49901
GNE myopathy	GNE	AR	Asian	1 in 58	99%	1 in 5701
GNE myopathy	GNE	AR	Sephardic Jewish - Iranian, Syrian	1 in 12	99%	1 in 1101
GRACILE syndrome	BCS1L	AR	Caucasian	1 in 407	99%	1 in 40601
GRACILE syndrome	BCS1L	AR	General Population	1 in 111	99%	1 in 11001
GRACILE syndrome	BCS1L	AR	Finnish	1 in 108	99%	1 in 10701
Guanidinoacetate methyltransferase deficiency	GAMT	AR	General Population	<1 in 500	99%	<1 in 49901
Guanidinoacetate methyltransferase deficiency	GAMT	AR	Portuguese	1 in 125	99%	1 in 12401
Hereditary fructose intolerance	ALDOB	AR	Hispanic	<1 in 500	99%	<1 in 49901
Hereditary fructose intolerance	ALDOB	AR	African American	1 in 406	99%	1 in 40501
Hereditary fructose intolerance	ALDOB	AR	Caucasian	1 in 80	99%	1 in 7901
Hereditary fructose intolerance	ALDOB	AR	General Population	1 in 55	99%	1 in 5401
Hereditary hemochromatosis, type 2	HJV	AR	Caucasian	<1 in 500	99%	<1 in 49901
Hereditary hemochromatosis, type 2	HJV	AR	General Population	<1 in 500	99%	<1 in 49901
Hereditary hemochromatosis, type 3	TFR2	AR	General Population	<1 in 500	99%	<1 in 49901
Hermansky-Pudlak syndrome, type 1	HPS1	AR	General Population	<1 in 500	99%	<1 in 49901
Hermansky-Pudlak syndrome, type 1	HPS1	AR	Puerto Rican	1 in 59	99%	1 in 5801
Hermansky-Pudlak syndrome, type 2	AP3B1	AR	General Population	1 in 158	99%	1 in 15701
Hermansky-Pudlak syndrome, type 3	HPS3	AR	General Population	<1 in 500	99%	<1 in 49901
Hermansky-Pudlak syndrome, type 3	HPS3	AR	Ashkenazi Jewish	1 in 235	99%	1 in 23401
Hermansky-Pudlak syndrome, type 4	HPS4	AR	General Population	<1 in 500	99%	<1 in 49901
Holocarboxylase synthetase deficiency	HLCS	AR	Caucasian	<1 in 500	99%	<1 in 49901
Holocarboxylase synthetase deficiency	HLCS	AR	General Population	<1 in 500	99%	<1 in 49901
Holocarboxylase synthetase deficiency	HLCS	AR	Asian	1 in 158	99%	1 in 15701
Holocarboxylase synthetase deficiency	HLCS	AR	Faroese	1 in 20	99%	1 in 1901
Homocystinuria caused by methylenetetrahydrofolate reductase (MTHFR) deficiency	MTHFR	AR	General Population	1 in 158	99%	1 in 15701
Homocystinuria caused by methylenetetrahydrofolate reductase (MTHFR) deficiency	MTHFR	AR	Sephardic Jewish - Bukharian	1 in 39	99%	1 in 3801
Homocystinuria, CBS-related	CBS	AR	General Population	1 in 224	99%	1 in 22301
Homocystinuria, CBS-related	CBS	AR	Caucasian	1 in 52	99%	1 in 5101
Homocystinuria, CBS-related	CBS	AR	Qatari	1 in 21	99%	1 in 2001
Homocystinuria, type cblE	MTRR	AR	General Population	<1 in 500	99%	<1 in 49901
Homocystinuria, type cblE	MTRR	AR	Caucasian	<1 in 500	99%	<1 in 49901
HPRT1-related disorders	HPRT1	XL	General Population	1 in 285000	99%	1 in 28499901
Hydrolethalus syndrome	HYLS1	AR	General Population	1 in 455	99%	1 in 45401
Hydrolethalus syndrome	HYLS1	AR	Finnish	1 in 50	99%	1 in 4901
Hyperphosphatemic familial tumoral calcinosis	GALNT3	AR	General Population	<1 in 500	99%	<1 in 49901
Hypohidrotic ectodermal dysplasia, X-linked	EDA	XL	General Population	1 in 3800	99%	1 in 379901
Hypophosphatasia	ALPL	AR	Northern European Caucasian	1 in 274	99%	1 in 27301
Hypophosphatasia	ALPL	AR	East Asian	1 in 203	99%	1 in 20201
Hypophosphatasia	ALPL	AR	Asian	1 in 203	99%	1 in 20201
Hypophosphatasia	ALPL	AR	General Population	1 in 158	99%	1 in 15701
Hypophosphatasia	ALPL	AR	Mennonite	1 in 25	99%	1 in 2401
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	FOXP3	XL	General Population	<1 in 750000	99%	<1 in 74999901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Infantile cerebral and cerebellar atrophy	MED17	AR	General Population	<1 in 500	99%	<1 in 49901
Infantile cerebral and cerebellar atrophy	MED17	AR	Sephardic Jewish - Bukharian, Kurdish	1 in 20	99%	1 in 1901
Isovaleric acidemia	IVD	AR	General Population	1 in 250	99%	1 in 24901
Isovaleric acidemia	IVD	AR	Caucasian	1 in 144	99%	1 in 14301
Isovaleric acidemia	IVD	AR	Asian	1 in 75	99%	1 in 7401
Johanson-Blizzard syndrome	UBR1	AR	General Population	1 in 250	99%	1 in 24901
Joubert syndrome 1	INPP5E	AR	General Population	1 in 264	99%	1 in 26301
Joubert syndrome 15	CEP41	AR	General Population	<1 in 500	99%	<1 in 49901
Joubert syndrome 17	CPLANE1	AR	General Population	1 in 423	99%	1 in 42201
Joubert syndrome 2	TMEM216	AR	General Population	<1 in 500	99%	<1 in 49901
Joubert syndrome 2	TMEM216	AR	Ashkenazi Jewish	1 in 110	99%	1 in 10901
Joubert syndrome 21	CSPP1	AR	General Population	<1 in 500	99%	<1 in 49901
Joubert syndrome 25	CEP104	AR	General Population	<1 in 500	99%	<1 in 49901
Joubert syndrome 27	B9D1	AR	General Population	<1 in 500	99%	<1 in 49901
Joubert syndrome 3	AHI1	AR	General Population	1 in 176	99%	1 in 17501
Joubert syndrome 31	CEP120	AR	General Population	<1 in 500	99%	<1 in 49901
Joubert syndrome 34	B9D2	AR	General Population	<1 in 500	99%	<1 in 49901
Joubert syndrome 8	ARL13B	AR	General Population	<1 in 500	99%	<1 in 49901
Joubert syndrome 9	CC2D2A	AR	General Population	<1 in 500	99%	<1 in 49901
Junctional epidermolysis bullosa, LAMA3-related	LAMA3	AR	General Population	<1 in 500	99%	<1 in 49901
Junctional epidermolysis bullosa, LAMB3-related	LAMB3	AR	General Population	1 in 407	99%	1 in 40601
Junctional epidermolysis bullosa, LAMC2-related	LAMC2	AR	General Population	<1 in 500	99%	<1 in 49901
Juvenile retinoschisis, X-linked	RS1	XL	General Population	1 in 2500	99%	1 in 249901
Krabbe disease	GALC	AR	Asian	<1 in 500	99%	<1 in 49901
Krabbe disease	GALC	AR	General Population	1 in 150	99%	1 in 14901
Krabbe disease	GALC	AR	Druze Northern Israel	1 in 6	99%	1 in 501
Krabbe disease	GALC	AR	Muslim Arab (Jerusalem)	1 in 6	99%	1 in 501
L1 syndrome	L1CAM	XL	General Population	1 in 22500	99%	1 in 2249901
LAMA2 muscular dystrophy	LAMA2	AR	General Population	1 in 87	99%	1 in 8601
Leber congenital amaurosis 13	RDH12	AR	General Population	1 in 456	99%	1 in 45501
Leber congenital amaurosis 2	RPE65	AR	General Population	1 in 228	99%	1 in 22701
Leber congenital amaurosis 2	RPE65	AR	Sephardic Jewish - North African	1 in 90	99%	1 in 8901
Leber congenital amaurosis 5	LCA5	AR	General Population	<1 in 500	99%	<1 in 49901
Leber congenital amaurosis, CEP290-related / CEP290-related conditions	CEP290	AR	General Population	1 in 185	99%	1 in 18401
Lethal congenital contracture syndrome 1	GLE1	AR	General Population	<1 in 500	99%	<1 in 49901
Lethal congenital contracture syndrome 1	GLE1	AR	Finnish	1 in 100	99%	1 in 9901
Leukoencephalopathy with vanishing white matter	EIF2B5	AR	General Population	<1 in 500	99%	<1 in 49901
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	General Population	<1 in 500	99%	<1 in 49901
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	Hispanic	1 in 260	99%	1 in 25901
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	East Asian	1 in 232	99%	1 in 23101
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	Northern European Caucasian	1 in 103	99%	1 in 10201
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	Caucasian	1 in 103	99%	1 in 10201
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	Amish	1 in 50	99%	1 in 4901
Limb-girdle muscular dystrophy, type 2B	DYSF	AR	General Population	1 in 311	99%	1 in 31001
Limb-girdle muscular dystrophy, type 2B	DYSF	AR	Caucasian	1 in 158	99%	1 in 15701
Limb-girdle muscular dystrophy, type 2B	DYSF	AR	Sephardic Jewish, Libyan, Moroccan, Tunisian, Bulgarian	1 in 14	99%	1 in 1301

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	FKRP	AR	General Population	1 in 158	99%	1 in 15701
Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	FKRP	AR	Norwegian	1 in 116	99%	1 in 11501
Limb-girdle muscular dystrophy, type 3	SGCA	AR	General Population	<1 in 500	99%	<1 in 49901
Limb-girdle muscular dystrophy, type 3	SGCA	AR	Caucasian	1 in 290	99%	1 in 28901
Limb-girdle muscular dystrophy, type 3	SGCA	AR	Northern European Caucasian	1 in 160	99%	1 in 15901
Limb-girdle muscular dystrophy, type 3	SGCA	AR	Finnish	1 in 150	99%	1 in 14901
Limb-girdle muscular dystrophy, type 4	SGCB	AR	Northern European Caucasian	<1 in 500	99%	<1 in 49901
Limb-girdle muscular dystrophy, type 4	SGCB	AR	General Population	<1 in 500	99%	<1 in 49901
Limb-girdle muscular dystrophy, type 4	SGCB	AR	Caucasian	1 in 406	99%	1 in 40501
Limb-girdle muscular dystrophy, type 5	SGCG	AR	Northern European Caucasian	<1 in 500	99%	<1 in 49901
Limb-girdle muscular dystrophy, type 5	SGCG	AR	General Population	1 in 354	99%	1 in 35301
Limb-girdle muscular dystrophy, type 5	SGCG	AR	Moroccan	1 in 250	99%	1 in 24901
Limb-girdle muscular dystrophy, type 5	SGCG	AR	Roma	1 in 96	99%	1 in 9501
Limb-girdle muscular dystrophy, type 6	SGCD	AR	General Population	<1 in 500	99%	<1 in 49901
Lipoid congenital adrenal hyperplasia	STAR	AR	General Population	<1 in 500	99%	<1 in 49901
Lipoid congenital adrenal hyperplasia	STAR	AR	East Asian	1 in 177	99%	1 in 17601
Lipoprotein lipase deficiency	LPL	AR	General Population	<1 in 500	99%	<1 in 49901
Lipoprotein lipase deficiency	LPL	AR	Caucasian	<1 in 500	99%	<1 in 49901
Lipoprotein lipase deficiency	LPL	AR	Asian	1 in 189	99%	1 in 18801
Lipoprotein lipase deficiency	LPL	AR	French Canadian - Other	1 in 139	99%	1 in 13801
Lipoprotein lipase deficiency	LPL	AR	French Canadian - Saguenay Lac-St. Jean	1 in 46	99%	1 in 4501
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA	AR	Caucasian	1 in 254	99%	1 in 25301
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA	AR	Finnish	1 in 240	99%	1 in 23901
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	HADHA	AR	General Population	1 in 138	99%	1 in 13701
Lowe syndrome, X-linked	OCRL	XL	General Population	1 in 375000	99%	1 in 37499901
Lysinuric protein intolerance	SLC7A7	AR	General Population	<1 in 500	99%	<1 in 49901
Lysinuric protein intolerance	SLC7A7	AR	Finnish	1 in 122	99%	1 in 12101
Lysinuric protein intolerance	SLC7A7	AR	Japanese	1 in 119	99%	1 in 11801
Lysosomal acid lipase deficiency	LIPA	AR	General Population	<1 in 500	99%	<1 in 49901
Lysosomal acid lipase deficiency	LIPA	AR	Ashkenazi Jewish	<1 in 500	99%	<1 in 49901
Lysosomal acid lipase deficiency	LIPA	AR	Caucasian	1 in 145	99%	1 in 14401
Lysosomal acid lipase deficiency	LIPA	AR	Sephardic Jewish - Iranian	1 in 26	99%	1 in 2501
Malonyl-CoA decarboxylase deficiency	MLYCD	AR	General Population	<1 in 500	99%	<1 in 49901
Maple syrup urine disease, type 1A	BCKDHA	AR	General Population	1 in 321	99%	1 in 32001
Maple syrup urine disease, type 1A	BCKDHA	AR	Caucasian	1 in 320	99%	1 in 31901
Maple syrup urine disease, type 1A	BCKDHA	AR	Portuguese Roma	1 in 71	99%	1 in 7001
Maple syrup urine disease, type 1A	BCKDHA	AR	Mennonite	1 in 10	99%	1 in 901
Maple syrup urine disease, type 1B	BCKDHB	AR	Caucasian	1 in 433	99%	1 in 43201
Maple syrup urine disease, type 1B	BCKDHB	AR	General Population	1 in 364	99%	1 in 36301
Maple syrup urine disease, type 1B	BCKDHB	AR	Asian	1 in 163	99%	1 in 16201
Maple syrup urine disease, type 1B	BCKDHB	AR	Ashkenazi Jewish	1 in 97	99%	1 in 9601
Maple syrup urine disease, type 2	DBT	AR	General Population	1 in 321	99%	1 in 32001
Medium chain acyl-CoA dehydrogenase deficiency	ACADM	AR	Asian	1 in 178	99%	1 in 17701
Medium chain acyl-CoA dehydrogenase deficiency	ACADM	AR	Caucasian	1 in 64	99%	1 in 6301
Medium chain acyl-CoA dehydrogenase deficiency	ACADM	AR	General Population	1 in 35	99%	1 in 3401
MEDNIK syndrome	AP1S1	AR	General Population	<1 in 500	99%	<1 in 49901
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	AR	General Population	<1 in 500	99%	<1 in 49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	AR	Libyan Jewish	1 in 40	99%	1 in 3901
Menkes disease	ATP7A	XL	General Population	1 in 26000	99%	1 in 2599901
Metachromatic leukodystrophy due to saposin B deficiency	PSAP	AR	General Population	<1 in 500	99%	<1 in 49901
Metachromatic leukodystrophy, ARSA-related	ARSA	AR	Ashkenazi Jewish	<1 in 500	99%	<1 in 49901
Metachromatic leukodystrophy, ARSA-related	ARSA	AR	General Population	1 in 100	99%	1 in 9901
Metachromatic leukodystrophy, ARSA-related	ARSA	AR	Sephardic Jewish - Yemenite	1 in 46	99%	1 in 4501
Metachromatic leukodystrophy, ARSA-related	ARSA	AR	Navajo	1 in 25	99%	1 in 2401
Methylmalonic aciduria, MMAA-related	MMAA	AR	General Population	1 in 316	99%	1 in 31501
Methylmalonic aciduria, MMAA-related	MMAA	AR	Caucasian	1 in 316	99%	1 in 31501
Methylmalonic aciduria, MMAB-related	MMAB	AR	Caucasian	1 in 456	99%	1 in 45501
Methylmalonic aciduria, MMAB-related	MMAB	AR	General Population	1 in 456	99%	1 in 45501
Methylmalonic aciduria, MMUT-related	MMUT	AR	General Population	1 in 383	99%	1 in 38201
Methylmalonic aciduria, MMUT-related	MMUT	AR	Hispanic	1 in 383	99%	1 in 38201
Methylmalonic aciduria, MMUT-related	MMUT	AR	Caucasian	1 in 224	99%	1 in 22301
Methylmalonic aciduria, MMUT-related	MMUT	AR	African American	1 in 177	99%	1 in 17601
Methylmalonic aciduria, MMUT-related	MMUT	AR	Asian	1 in 53	99%	1 in 5201
Mevalonic aciduria / Hyper-IgD syndrome	MVK	AR	General Population	1 in 167	99%	1 in 16601
Micropthalmia / Anopthalmia	VSX2	AR	General Population	<1 in 500	99%	<1 in 49901
Micropthalmia / Anopthalmia	VSX2	AR	Sephardic Jewish - Iranian, Syrian	1 in 145	99%	1 in 14401
Mitochondrial complex I deficiency, ACAD9-related	ACAD9	AR	General Population	<1 in 500	99%	<1 in 49901
Mitochondrial complex I deficiency, nuclear type 1	NDUFS4	AR	General Population	1 in 423	99%	1 in 42201
Mitochondrial complex I deficiency, nuclear type 16	NDUFAF5	AR	General Population	<1 in 500	99%	<1 in 49901
Mitochondrial complex I deficiency, nuclear type 16	NDUFAF5	AR	Ashkenazi Jewish	1 in 290	99%	1 in 28901
Mitochondrial complex I deficiency, nuclear type 17	NDUFAF6	AR	General Population	<1 in 500	99%	<1 in 49901
Mitochondrial complex I deficiency, nuclear type 9	NDUFS6	AR	General Population	<1 in 500	99%	<1 in 49901
Mitochondrial complex I deficiency, nuclear type 9	NDUFS6	AR	Caucasus Jewish	1 in 24	99%	1 in 2301
Mitochondrial complex IV deficiency, nuclear type 12	PET100	AR	General Population	1 in 452	99%	1 in 45101
Mitochondrial complex IV deficiency, nuclear type 2	SCO2	AR	General Population	1 in 149	99%	1 in 14801
Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type	LRPPRC	AR	General Population	<1 in 500	99%	<1 in 49901
Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type	LRPPRC	AR	French Canadian - Saguenay Lac-St. Jean	1 in 23	99%	1 in 2201
Mitochondrial DNA depletion syndrome 1, MNGIE type	TYMP	AR	Caucasian	<1 in 500	99%	<1 in 49901
Mitochondrial DNA depletion syndrome 1, MNGIE type	TYMP	AR	General Population	<1 in 500	99%	<1 in 49901
Mitochondrial DNA depletion syndrome 1, MNGIE type	TYMP	AR	Sephardic Jewish - Iranian	1 in 158	99%	1 in 15701
Mitochondrial trifunctional protein deficiency, HADHB-related	HADHB	AR	General Population	1 in 146	99%	1 in 14501
MKS1-related disorders	MKS1	AR	General Population	1 in 260	99%	1 in 25901
MKS1-related disorders	MKS1	AR	Caucasian	1 in 260	99%	1 in 25901
MKS1-related disorders	MKS1	AR	Finnish	1 in 47	99%	1 in 4601
Molybdenum cofactor deficiency of complementation group A	MOCS1	AR	General Population	<1 in 500	99%	<1 in 49901
MPV17-related mitochondrial DNA (mtDNA) maintenance defect	MPV17	AR	General Population	<1 in 500	99%	<1 in 49901
MPV17-related mitochondrial DNA (mtDNA) maintenance defect	MPV17	AR	Navajo	1 in 20	99%	1 in 1901
Mucolipidosis II and mucolipidosis III alpha/beta	GNPTAB	AR	Asian	1 in 389	99%	1 in 38801
Mucolipidosis II and mucolipidosis III alpha/beta	GNPTAB	AR	Caucasian	1 in 225	99%	1 in 22401
Mucolipidosis II and mucolipidosis III alpha/beta	GNPTAB	AR	General Population	1 in 158	99%	1 in 15701
Mucolipidosis III gamma	GNPTG	AR	General Population	<1 in 500	99%	<1 in 49901
Mucolipidosis III gamma	GNPTG	AR	Caucasian	1 in 273	99%	1 in 27201

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Mucolipidosis, type IV	MCOLN1	AR	General Population	<1 in 500	99%	<1 in 49901
Mucolipidosis, type IV	MCOLN1	AR	Ashkenazi Jewish	1 in 89	99%	1 in 8801
Mucopolysaccharidosis, type I / Hurler syndrome	IDUA	AR	General Population	1 in 158	99%	1 in 15701
Mucopolysaccharidosis, type I / Hurler syndrome	IDUA	AR	Northern European Caucasian	1 in 145	99%	1 in 14401
Mucopolysaccharidosis, type II / Hunter syndrome	IDS	XL	General Population	1 in 60000	90%	1 in 599991
Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A	SGSH	AR	General Population	1 in 415	99%	1 in 41401
Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A	SGSH	AR	Caucasian	1 in 253	99%	1 in 25201
Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	NAGLU	AR	General Population	<1 in 500	99%	<1 in 49901
Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	NAGLU	AR	Caucasian	1 in 346	99%	1 in 34501
Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	NAGLU	AR	Asian	1 in 298	99%	1 in 29701
Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	HGSNAT	AR	Asian	<1 in 500	99%	<1 in 49901
Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	HGSNAT	AR	General Population	1 in 482	99%	1 in 48101
Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	HGSNAT	AR	Caucasian	1 in 259	99%	1 in 25801
Mucopolysaccharidosis, type IIID / Sanfilippo syndrome D	GNS	AR	General Population	<1 in 500	99%	<1 in 49901
Mucopolysaccharidosis, type IVA / Morquio syndrome	GALNS	AR	General Population	1 in 307	99%	1 in 30601
Mucopolysaccharidosis, type IX / Hyaluronidase deficiency	HYAL1	AR	General Population	<1 in 500	99%	<1 in 49901
Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	ARSB	AR	Asian	1 in 423	99%	1 in 42201
Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	ARSB	AR	General Population	1 in 291	99%	1 in 29001
Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	ARSB	AR	Caucasian	1 in 273	99%	1 in 27201
Mucopolysaccharidosis, type VII / Sly syndrome	GUSB	AR	General Population	<1 in 500	99%	<1 in 49901
Mulibrey nanism	TRIM37	AR	General Population	<1 in 500	99%	<1 in 49901
Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIA	ETFA	AR	General Population	<1 in 500	99%	<1 in 49901
Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIB	ETFB	AR	General Population	1 in 408	99%	1 in 40701
Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIC	ETFDH	AR	General Population	1 in 250	99%	1 in 24901
Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIC	ETFDH	AR	Asian	1 in 87	99%	1 in 8601
Multiple pterygium syndrome, lethal type	CHRNG	AR	General Population	1 in 50	99%	1 in 4901
Multiple sulfatase deficiency	SUMF1	AR	General Population	<1 in 500	99%	<1 in 49901
Multiple sulfatase deficiency	SUMF1	AR	Ashkenazi Jewish	1 in 279	99%	1 in 27801
Muscular dystrophy-dystroglycanopathy, type A, 1	POMT1	AR	General Population	1 in 275	99%	1 in 27401
Muscular dystrophy-dystroglycanopathy, type A, 2	POMT2	AR	General Population	1 in 465	99%	1 in 46401
Muscular dystrophy-dystroglycanopathy, type A, 3	POMGNT1	AR	General Population	1 in 462	99%	1 in 46101
Muscular dystrophy-dystroglycanopathy, type A, 3	POMGNT1	AR	Finnish	1 in 111	99%	1 in 11001
Muscular dystrophy-dystroglycanopathy, type A, 6	LARGE1	AR	General Population	<1 in 500	99%	<1 in 49901
Muscular dystrophy-dystroglycanopathy, type A, 7	CRPPA	AR	General Population	1 in 371	99%	1 in 37001
Myopathy, lactic acidosis, and sideroblastic anemia	PUS1	AR	Sephardic Jewish - Iranian	<1 in 500	99%	<1 in 49901
Myopathy, lactic acidosis, and sideroblastic anemia	PUS1	AR	General Population	<1 in 500	99%	<1 in 49901
Myotonia congenita	CLCN1	AR	General Population	1 in 158	99%	1 in 15701
Myotonia congenita	CLCN1	AR	Finnish	1 in 59	99%	1 in 5801
Myotonia congenita	CLCN1	AR	Norwegian	1 in 53	99%	1 in 5201
N-acetylglutamate synthase deficiency	NAGS	AR	General Population	<1 in 500	99%	<1 in 49901
Nemaline myopathy 2	NEB	AR	General Population	1 in 224	95%	1 in 4461
Nemaline myopathy 2	NEB	AR	Ashkenazi Jewish	1 in 168	95%	1 in 3341
Nemaline myopathy 2	NEB	AR	Finnish	1 in 112	95%	1 in 2221
Nephrogenic diabetes insipidus	AQP2	AR	General Population	<1 in 500	99%	<1 in 49901
Nephronophthisis 2	INVS	AR	General Population	1 in 373	99%	1 in 37201
Neuronal ceroid lipofuscinosis, CLN3-related	CLN3	AR	Caucasian	1 in 188	99%	1 in 18701

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Neuronal ceroid lipofuscinosis, CLN3-related	CLN3	AR	General Population	1 in 145	99%	1 in 14401
Neuronal ceroid lipofuscinosis, CLN5-related	CLN5	AR	General Population	1 in 317	99%	1 in 31601
Neuronal ceroid lipofuscinosis, CLN5-related	CLN5	AR	Finnish	1 in 289	99%	1 in 28801
Neuronal ceroid lipofuscinosis, CLN6-related	CLN6	AR	General Population	1 in 261	99%	1 in 26001
Neuronal ceroid lipofuscinosis, CLN8-related	CLN8	AR	General Population	1 in 349	99%	1 in 34801
Neuronal ceroid lipofuscinosis, CLN8-related	CLN8	AR	Finnish	1 in 135	99%	1 in 13401
Neuronal ceroid lipofuscinosis, CTSD-related	CTSD	AR	General Population	<1 in 500	99%	<1 in 49901
Neuronal ceroid lipofuscinosis, MFSD8-related	MFSD8	AR	General Population	<1 in 500	99%	<1 in 49901
Neuronal ceroid lipofuscinosis, PPT1-related	PPT1	AR	General Population	1 in 368	99%	1 in 36701
Neuronal ceroid lipofuscinosis, PPT1-related	PPT1	AR	Finnish	1 in 70	99%	1 in 6901
Neuronal ceroid lipofuscinosis, TPP1-related	TPP1	AR	General Population	1 in 314	99%	1 in 31301
Neuronal ceroid lipofuscinosis, TPP1-related	TPP1	AR	Newfoundland	1 in 59	99%	1 in 5801
Niemann-Pick disease, type C1	NPC1	AR	Asian	1 in 404	99%	1 in 40301
Niemann-Pick disease, type C1	NPC1	AR	General Population	1 in 282	99%	1 in 28101
Niemann-Pick disease, type C1	NPC1	AR	Caucasian	1 in 185	99%	1 in 18401
Niemann-Pick disease, type C2	NPC2	AR	General Population	<1 in 500	99%	<1 in 49901
Niemann-Pick disease, types A/B	SMPD1	AR	Caucasian	1 in 244	99%	1 in 24301
Niemann-Pick disease, types A/B	SMPD1	AR	General Population	1 in 196	99%	1 in 19501
Niemann-Pick disease, types A/B	SMPD1	AR	Ashkenazi Jewish	1 in 115	99%	1 in 11401
Nijmegen breakage syndrome	NBN	AR	General Population	<1 in 500	99%	<1 in 49901
Nijmegen breakage syndrome	NBN	AR	Caucasian	1 in 155	99%	1 in 15401
Nonsyndromic hearing loss and deafness (DFNB) 1	GJB2	AR	General Population	1 in 42	99%	1 in 4101
Nonsyndromic hearing loss and deafness (DFNB) 1	GJB2	AR	Caucasian	1 in 30	99%	1 in 2901
Nonsyndromic hearing loss and deafness (DFNB) 1	GJB2	AR	Ashkenazi Jewish	1 in 21	99%	1 in 2001
Nonsyndromic hearing loss and deafness (DFNB) 1	GJB2	AR	East Asian	1 in 10	99%	1 in 901
Nonsyndromic hearing loss and deafness (DFNB) 3	MYO15A	AR	General Population	1 in 117	99%	1 in 11601
Nonsyndromic hearing loss and deafness (DFNB) 77	LOXHD1	AR	General Population	<1 in 500	99%	<1 in 49901
Nonsyndromic hearing loss and deafness (DFNB) 77	LOXHD1	AR	Ashkenazi Jewish	1 in 180	99%	1 in 17901
NPHP1 nephronophthisis-related ciliopathies	NPHP1	AR	General Population	1 in 202	99%	1 in 20101
NPHP3 nephronophthisis-related ciliopathies	NPHP3	AR	General Population	<1 in 500	99%	<1 in 49901
NPHP4 nephronophthisis-related ciliopathies	NPHP4	AR	General Population	<1 in 500	99%	<1 in 49901
Oculocutaneous albinism, type I	TYR	AR	General Population	1 in 20	99%	1 in 1901
Oculocutaneous albinism, type II	OCA2	AR	General Population	1 in 76	99%	1 in 7501
Oculocutaneous albinism, type II	OCA2	AR	African American	1 in 50	99%	1 in 4901
Oculocutaneous albinism, type II	OCA2	AR	Navajo	1 in 22	99%	1 in 2101
Odonto-onycho-dermal dysplasia / Schopf-Schulz-Passarge syndrome	WNT10A	AR	General Population	1 in 305	99%	1 in 30401
Omenn syndrome	DCLRE1C	AR	Northern European Caucasian	<1 in 500	99%	<1 in 49901
Omenn syndrome	DCLRE1C	AR	General Population	<1 in 500	99%	<1 in 49901
Omenn syndrome	DCLRE1C	AR	Navajo and Apache Native Americans	1 in 10	99%	1 in 901
Ornithine aminotransferase deficiency	OAT	AR	General Population	<1 in 500	99%	<1 in 49901
Ornithine aminotransferase deficiency	OAT	AR	Sephardic Jewish - Iraqi, Syrian	1 in 177	99%	1 in 17601
Ornithine aminotransferase deficiency	OAT	AR	Finnish	1 in 147	99%	1 in 14601
Ornithine transcarbamylase deficiency, X-linked	OTC	XL	General Population	1 in 30000	99%	1 in 2999901
Ornithine translocase deficiency	SLC25A15	AR	General Population	<1 in 500	99%	<1 in 49901
Ornithine translocase deficiency	SLC25A15	AR	French Canadian	1 in 20	99%	1 in 1901
Ornithine translocase deficiency	SLC25A15	AR	Metis from Saskatchewan	1 in 19	99%	1 in 1801
Orofaciodigital syndrome, type XIV	C2CD3	AR	General Population	<1 in 500	99%	<1 in 49901
Osteopetrosis, infantile malignant, TCIRG1-related	TCIRG1	AR	Ashkenazi Jewish	1 in 350	99%	1 in 34901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Osteopetrosis, infantile malignant, TCIRG1-related	TCIRG1	AR	General Population	1 in 316	99%	1 in 31501
Osteopetrosis, infantile malignant, TCIRG1-related	TCIRG1	AR	Costa Rican	1 in 86	99%	1 in 8501
Osteopetrosis, infantile malignant, TCIRG1-related	TCIRG1	AR	Chuvashiya	1 in 60	99%	1 in 5901
Pendred syndrome	SLC26A4	AR	Caucasian	1 in 88	99%	1 in 8701
Pendred syndrome	SLC26A4	AR	General Population	1 in 80	99%	1 in 7901
Pendred syndrome	SLC26A4	AR	African American	1 in 76	99%	1 in 7501
Pendred syndrome	SLC26A4	AR	Asian	1 in 74	99%	1 in 7301
Pendred syndrome	SLC26A4	AR	Northern European Caucasian	1 in 60	99%	1 in 5901
Perlman syndrome	DIS3L2	AR	General Population	<1 in 500	99%	<1 in 49901
Peroxisomal acyl-CoA oxidase deficiency	ACOX1	AR	General Population	<1 in 500	99%	<1 in 49901
Phenylalanine hydroxylase deficiency	PAH	AR	Finnish	1 in 225	99%	1 in 22401
Phenylalanine hydroxylase deficiency	PAH	AR	Ashkenazi Jewish	1 in 225	99%	1 in 22401
Phenylalanine hydroxylase deficiency	PAH	AR	Hispanic American	1 in 163	99%	1 in 16201
Phenylalanine hydroxylase deficiency	PAH	AR	African American	1 in 143	99%	1 in 14201
Phenylalanine hydroxylase deficiency	PAH	AR	Asian	1 in 78	99%	1 in 7701
Phenylalanine hydroxylase deficiency	PAH	AR	General Population	1 in 65	99%	1 in 6401
Phenylalanine hydroxylase deficiency	PAH	AR	Caucasian	1 in 50	99%	1 in 4901
Phenylalanine hydroxylase deficiency	PAH	AR	Southern European Caucasian	1 in 40	99%	1 in 3901
Phenylalanine hydroxylase deficiency	PAH	AR	Irish	1 in 34	99%	1 in 3301
Phenylalanine hydroxylase deficiency	PAH	AR	Turkish	1 in 32	99%	1 in 3101
Phenylalanine hydroxylase deficiency	PAH	AR	Sicilian	1 in 26	99%	1 in 2501
Phenylalanine hydroxylase deficiency	PAH	AR	Sephardic Jewish - Iranian, Bukharian, Kavkazi, Tunisian, Moroccan	1 in 18	99%	1 in 1701
Phosphoglycerate dehydrogenase deficiency	PHGDH	AR	General Population	<1 in 500	99%	<1 in 49901
Phosphoglycerate dehydrogenase deficiency	PHGDH	AR	Ashkenazi Jewish	1 in 453	99%	1 in 45201
PLA2G6-associated neurodegeneration	PLA2G6	AR	General Population	<1 in 500	99%	<1 in 49901
PLP1-related disorders	PLP1	XL	Northern European Caucasian	1 in 57700	99%	1 in 5769901
PLP1-related disorders	PLP1	XL	General Population	1 in 37500	99%	1 in 3749901
POLG-related disorders	POLG	AR	General Population	1 in 50	99%	1 in 4901
Pontocerebellar hypoplasia, type 1A	VRK1	AR	General Population	<1 in 500	99%	<1 in 49901
Pontocerebellar hypoplasia, type 1A	VRK1	AR	Ashkenazi Jewish	1 in 225	99%	1 in 22401
Pontocerebellar hypoplasia, type 1B	EXOSC3	AR	General Population	<1 in 500	99%	<1 in 49901
Pontocerebellar hypoplasia, type 2B	TSEN2	AR	General Population	<1 in 500	99%	<1 in 49901
Pontocerebellar hypoplasia, type 2E	VPS53	AR	General Population	<1 in 500	99%	<1 in 49901
Pontocerebellar hypoplasia, type 6	RARS2	AR	General Population	<1 in 500	99%	<1 in 49901
Pontocerebellar hypoplasia, type 6	RARS2	AR	Sephardic Jewish - Iraqi, Syrian, Tunisian	<1 in 500	99%	<1 in 49901
Pontocerebellar hypoplasia, types 4 and 2A	TSEN54	AR	General Population	<1 in 500	99%	<1 in 49901
Primary ciliary dyskinesia, DNAH5-related	DNAH5	AR	Ashkenazi Jewish	1 in 174	99%	1 in 17301
Primary ciliary dyskinesia, DNAH5-related	DNAH5	AR	General Population	1 in 120	99%	1 in 11901
Primary ciliary dyskinesia, DNAI1-related	DNAI1	AR	Ashkenazi Jewish	1 in 352	99%	1 in 35101
Primary ciliary dyskinesia, DNAI1-related	DNAI1	AR	General Population	1 in 182	99%	1 in 18101
Primary ciliary dyskinesia, DNAI2-related	DNAI2	AR	General Population	<1 in 500	99%	<1 in 49901
Primary ciliary dyskinesia, DNAI2-related	DNAI2	AR	Ashkenazi Jewish	1 in 200	99%	1 in 19901
Primary congenital glaucoma	CYP1B1	AR	General Population	1 in 74	99%	1 in 7301
Primary hyperoxaluria, type I	AGXT	AR	General Population	1 in 158	99%	1 in 15701
Primary hyperoxaluria, type II	GRHPR	AR	General Population	<1 in 500	99%	<1 in 49901
Primary hyperoxaluria, type III	HOGA1	AR	General Population	1 in 309	99%	1 in 30801
Progressive cerebello-cerebral atrophy	SEPSECS	AR	General Population	<1 in 500	99%	<1 in 49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Progressive cerebello-cerebral atrophy	SEPSECS	AR	Sephardic Jewish - Moroccan, Iraqi	1 in 41	99%	1 in 4001
Progressive familial intrahepatic cholestasis 1 and benign familial intrahepatic cholestasis 1	ATP8B1	AR	General Population	1 in 53	99%	1 in 5201
Progressive familial intrahepatic cholestasis 4	TJP2	AR	General Population	<1 in 500	99%	<1 in 49901
Progressive familial intrahepatic cholestastasis, type 2	ABCB11	AR	General Population	1 in 158	99%	1 in 15701
Prolidase deficiency	PEPD	AR	General Population	1 in 242	99%	1 in 24101
Propionic acidemia, PCCA-related	PCCA	AR	Caucasian	1 in 380	99%	1 in 37901
Propionic acidemia, PCCA-related	PCCA	AR	General Population	1 in 224	99%	1 in 22301
Propionic acidemia, PCCA-related	PCCA	AR	Asian	1 in 162	99%	1 in 16101
Propionic acidemia, PCCB-related	PCCB	AR	General Population	1 in 224	99%	1 in 22301
Propionic acidemia, PCCB-related	PCCB	AR	Caucasian	1 in 202	99%	1 in 20101
Propionic acidemia, PCCB-related	PCCB	AR	Asian	1 in 145	99%	1 in 14401
PRPS1-related disorders	PRPS1	XL	General Population	1 in 500000	99%	1 in 49999901
Pseudocholinesterase deficiency	BCHE	AR	General Population	1 in 53	99%	1 in 5201
Pseudoxanthoma elasticum	ABCC6	AR	General Population	1 in 79	90%	1 in 781
Pycnodysostosis	CTSK	AR	General Population	1 in 439	99%	1 in 43801
Pyridoxine-dependent epilepsy	ALDH7A1	AR	General Population	1 in 158	99%	1 in 15701
Pyruvate carboxylase deficiency	PC	AR	General Population	1 in 250	99%	1 in 24901
Pyruvate dehydrogenase E1-alpha deficiency	PDHA1	XL	General Population	<1 in 750000	99%	<1 in 74999901
Pyruvate dehydrogenase E1-beta deficiency	PDHB	AR	General Population	<1 in 500	99%	<1 in 49901
Recurrent metabolic crises with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	TANGO2	AR	General Population	<1 in 500	99%	<1 in 49901
Refsum disease	PHYH	AR	General Population	<1 in 500	99%	<1 in 49901
Renal tubular acidosis	SLC4A4	AR	General Population	<1 in 500	99%	<1 in 49901
Renal tubular acidosis and deafness, ATP6V1B1-related	ATP6V1B1	AR	General Population	<1 in 500	99%	<1 in 49901
Renal tubular acidosis and deafness, ATP6V1B1-related	ATP6V1B1	AR	Sephardic Jewish - Syrian	1 in 140	99%	1 in 13901
Retinitis pigmentosa 25	EYS	AR	Ashkenazi Jewish	<1 in 500	99%	<1 in 49901
Retinitis pigmentosa 25	EYS	AR	General Population	1 in 129	99%	1 in 12801
Retinitis pigmentosa 25	EYS	AR	Caucasian	1 in 53	99%	1 in 5201
Retinitis pigmentosa 25	EYS	AR	Sephardic Jewish - Moroccan	1 in 42	99%	1 in 4101
Retinitis pigmentosa 26	CERKL	AR	General Population	1 in 137	99%	1 in 13601
Retinitis pigmentosa 26	CERKL	AR	Sephardic Jewish - Yemenite	1 in 24	99%	1 in 2301
Retinitis pigmentosa 28	FAM161A	AR	General Population	1 in 289	99%	1 in 28801
Retinitis pigmentosa 28	FAM161A	AR	Ashkenazi Jewish	1 in 214	99%	1 in 21301
Retinitis pigmentosa 28	FAM161A	AR	Sephardic Jewish, Libyan, Moroccan, Tunisian, Bulgarian	1 in 41	99%	1 in 4001
Retinitis pigmentosa 3	RPGR	XL	General Population	1 in 20000	99%	1 in 1999901
Retinitis pigmentosa 59	DHDDS	AR	General Population	<1 in 500	99%	<1 in 49901
Retinitis pigmentosa 59	DHDDS	AR	Ashkenazi Jewish	1 in 117	99%	1 in 11601
Rhizomelic chondrodysplasia punctata, type 1	PEX7	AR	General Population	<1 in 500	99%	<1 in 49901
Rhizomelic chondrodysplasia punctata, type 1	PEX7	AR	Caucasian	1 in 158	99%	1 in 15701
Rhizomelic chondrodysplasia punctata, type 2	GNPAT	AR	General Population	<1 in 500	99%	<1 in 49901
Rhizomelic chondrodysplasia punctata, type 3	AGPS	AR	General Population	<1 in 500	99%	<1 in 49901
Roberts-SC phocomelia syndrome	ESCO2	AR	General Population	<1 in 500	99%	<1 in 49901
Sandhoff disease	HEXB	AR	Ashkenazi Jewish	<1 in 500	99%	<1 in 49901
Sandhoff disease	HEXB	AR	General Population	1 in 278	99%	1 in 27701
Sandhoff disease	HEXB	AR	Caucasian	1 in 235	99%	1 in 23401
Sandhoff disease	HEXB	AR	Argentinian Creole	1 in 64	99%	1 in 6301
Schimke immunoosseous dysplasia	SMARCAL1	AR	General Population	<1 in 500	99%	<1 in 49901
Schindler disease	NAGA	AR	General Population	1 in 94	99%	1 in 9301

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Senior-Loken syndrome 5	IQCB1	AR	General Population	<1 in 500	99%	<1 in 49901
Severe combined immunodeficiency, RAG1-related	RAG1	AR	General Population	1 in 245	99%	1 in 24401
Severe combined immunodeficiency, RAG2-related	RAG2	AR	General Population	<1 in 500	99%	<1 in 49901
Severe combined immunodeficiency, RAG2-related	RAG2	AR	Sephardic Jewish - Iraqi	<1 in 500	99%	<1 in 49901
Severe combined immunodeficiency, X-linked	IL2RG	XL	General Population	1 in 69000	99%	1 in 6899901
Severe congenital neutropenia 5	VPS45	AR	General Population	<1 in 500	99%	<1 in 49901
Short-rib thoracic dysplasia 3 with or without polydactyly	DYNC2H1	AR	East Asian	<1 in 500	99%	<1 in 49901
Short-rib thoracic dysplasia 3 with or without polydactyly	DYNC2H1	AR	Japanese	1 in 297	99%	1 in 29601
Short-rib thoracic dysplasia 3 with or without polydactyly	DYNC2H1	AR	General Population	1 in 67	99%	1 in 6601
Shwachman-Diamond syndrome	SBDS	AR	General Population	1 in 145	99%	1 in 14401
Sialidosis	NEU1	AR	General Population	<1 in 500	99%	<1 in 49901
Sjogren-Larsson syndrome	ALDH3A2	AR	Northern European Caucasian	1 in 223	99%	1 in 22201
Sjogren-Larsson syndrome	ALDH3A2	AR	General Population	1 in 223	99%	1 in 22201
Sjogren-Larsson syndrome	ALDH3A2	AR	Swedish	1 in 204	99%	1 in 20301
Skeletal dysplasias, SLC26A2-related	SLC26A2	AR	General Population	1 in 158	99%	1 in 15701
Skeletal dysplasias, SLC26A2-related	SLC26A2	AR	Finnish	1 in 50	99%	1 in 4901
Smith-Lemli-Opitz syndrome	DHCR7	AR	Asian	<1 in 500	99%	<1 in 49901
Smith-Lemli-Opitz syndrome	DHCR7	AR	General Population	1 in 100	99%	1 in 9901
Smith-Lemli-Opitz syndrome	DHCR7	AR	African American	1 in 93	99%	1 in 9201
Smith-Lemli-Opitz syndrome	DHCR7	AR	Northern European Caucasian	1 in 50	99%	1 in 4901
Smith-Lemli-Opitz syndrome	DHCR7	AR	Caucasian	1 in 50	99%	1 in 4901
Smith-Lemli-Opitz syndrome	DHCR7	AR	Ashkenazi Jewish	1 in 36	99%	1 in 3501
Smith-Lemli-Opitz syndrome	DHCR7	AR	Ashkenazi Jewish	1 in 36	99%	1 in 3501
Spastic ataxia, Charlevoix-Saguenay type	SACS	AR	General Population	<1 in 500	99%	<1 in 49901
Spastic ataxia, Charlevoix-Saguenay type	SACS	AR	Caucasian	1 in 450	99%	1 in 44901
Spastic ataxia, Charlevoix-Saguenay type	SACS	AR	French Canadian - Charlevoix-Saguenay	1 in 21	99%	1 in 2001
Spastic paraplegia, type 15	ZFYVE26	AR	General Population	<1 in 500	99%	<1 in 49901
Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	SLC1A4	AR	General Population	<1 in 500	99%	<1 in 49901
Spinal muscular atrophy	SMN1	AR	Hispanic	1 in 117	99%	1 in 11601
Spinal muscular atrophy	SMN1	AR	African American	1 in 72	99%	1 in 7101
Spinal muscular atrophy	SMN1	AR	Hispanic American	1 in 68	99%	1 in 6701
Spinal muscular atrophy	SMN1	AR	Ashkenazi Jewish	1 in 67	99%	1 in 6601
Spinal muscular atrophy	SMN1	AR	East Asian	1 in 59	99%	1 in 5801
Spinal muscular atrophy	SMN1	AR	South Asian	1 in 59	99%	1 in 5801
Spinal muscular atrophy	SMN1	AR	Southeast Asian	1 in 59	99%	1 in 5801
Spinal muscular atrophy	SMN1	AR	Asian	1 in 59	99%	1 in 5801
Spinal muscular atrophy	SMN1	AR	General Population	1 in 54	99%	1 in 5301
Spinal muscular atrophy	SMN1	AR	Northern European Caucasian	1 in 47	99%	1 in 4601
Spinal muscular atrophy	SMN1	AR	Caucasian	1 in 47	99%	1 in 4601
Spondylothoracic dysostosis and spondylocostal dysostosis 2	MESP2	AR	General Population	1 in 224	99%	1 in 22301
Spondylothoracic dysostosis and spondylocostal dysostosis 2	MESP2	AR	Puerto Rican	1 in 55	99%	1 in 5401
Steel syndrome	COL27A1	AR	General Population	<1 in 500	99%	<1 in 49901
Steroid resistant nephrotic syndrome, type 1	NPHS1	AR	General Population	1 in 325	99%	1 in 32401
Steroid resistant nephrotic syndrome, type 1	NPHS1	AR	Finnish	1 in 45	99%	1 in 4401
Steroid resistant nephrotic syndrome, type 1	NPHS1	AR	Groffdale Conference Mennonites	1 in 12	99%	1 in 1101
Steroid-resistant nephrotic syndrome, type 2	NPHS2	AR	General Population	1 in 377	99%	1 in 37601
Steroid-resistant nephrotic syndrome, type 3	PLCE1	AR	General Population	<1 in 500	99%	<1 in 49901

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Stuve-Wiedemann syndrome	LIFR	AR	General Population	<1 in 500	99%	<1 in 49901
Surfactant dysfunction, ABCA3-related	ABCA3	AR	General Population	1 in 116	99%	1 in 11501
Surfactant dysfunction, ABCA3-related	ABCA3	AR	African American	1 in 68	99%	1 in 6701
Surfactant dysfunction, ABCA3-related	ABCA3	AR	Caucasian	1 in 28	99%	1 in 2701
Tay-Sachs disease	HEXA	AR	African American	1 in 271	99%	1 in 27001
Tay-Sachs disease	HEXA	AR	General Population	1 in 250	99%	1 in 24901
Tay-Sachs disease	HEXA	AR	Caucasian	1 in 182	99%	1 in 18101
Tay-Sachs disease	HEXA	AR	Asian	1 in 126	99%	1 in 12501
Tay-Sachs disease	HEXA	AR	Sephardic Jewish - Moroccan, Iraqi	1 in 125	99%	1 in 12401
Tay-Sachs disease	HEXA	AR	French Canadian - Other	1 in 73	99%	1 in 7201
Tay-Sachs disease	HEXA	AR	Irish	1 in 41	99%	1 in 4001
Tay-Sachs disease	HEXA	AR	Ashkenazi Jewish	1 in 27	99%	1 in 2601
Tay-Sachs disease	HEXA	AR	French Canadian	1 in 13	99%	1 in 1201
Tay-Sachs disease	HEXA	AR	French Canadian - Gaspesie	1 in 13	99%	1 in 1201
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	TECPR2	AR	General Population	<1 in 500	99%	<1 in 49901
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	TECPR2	AR	Sephardic Jewish - Bukharian	1 in 27	99%	1 in 2601
TNXB-related classical-like Ehlers-Danlos syndrome	TNXB	AR	General Population	1 in 28	95%	1 in 541
Trichohepatoenteric syndrome 1	TTC37	AR	General Population	1 in 381	99%	1 in 38001
Trimethylaminuria	FMO3	AR	General Population	1 in 139	99%	1 in 13801
Trimethylaminuria	FMO3	AR	Northern European Caucasian	1 in 100	99%	1 in 9901
Triple A syndrome	AAAS	AR	General Population	<1 in 500	99%	<1 in 49901
Tyrosine hydroxylase deficiency	TH	AR	General Population	<1 in 500	99%	<1 in 49901
Tyrosine hydroxylase deficiency	TH	AR	Asian	1 in 416	99%	1 in 41501
Tyrosine hydroxylase deficiency	TH	AR	Caucasian	1 in 224	99%	1 in 22301
Tyrosinemia, type I	FAH	AR	Asian	<1 in 500	99%	<1 in 49901
Tyrosinemia, type I	FAH	AR	African American	1 in 478	99%	1 in 47701
Tyrosinemia, type I	FAH	AR	Caucasian	1 in 333	99%	1 in 33201
Tyrosinemia, type I	FAH	AR	Ashkenazi Jewish	1 in 150	99%	1 in 14901
Tyrosinemia, type I	FAH	AR	General Population	1 in 100	99%	1 in 9901
Tyrosinemia, type I	FAH	AR	French Canadian (Qubec	1 in 66	99%	1 in 6501
Tyrosinemia, type I	FAH	AR	French Canadian - Saguenay Lac-St. Jean	1 in 25	99%	1 in 2401
Tyrosinemia, type II	TAT	AR	General Population	<1 in 500	99%	<1 in 49901
Usher syndrome, type 1B	MYO7A	AR	African American	<1 in 500	99%	<1 in 49901
Usher syndrome, type 1B	MYO7A	AR	General Population	1 in 206	99%	1 in 20501
Usher syndrome, type 1B	MYO7A	AR	Caucasian	1 in 145	99%	1 in 14401
Usher syndrome, type 1B	MYO7A	AR	Japanese	1 in 123	99%	1 in 12201
Usher syndrome, type 1B	MYO7A	AR	Asian	1 in 62	99%	1 in 6101
Usher syndrome, type 1C	USH1C	AR	General Population	1 in 353	99%	1 in 35201
Usher syndrome, type 1C	USH1C	AR	French Canadian / Acadian	1 in 227	99%	1 in 22601
Usher syndrome, type 1C	USH1C	AR	Acadian	1 in 41	99%	1 in 4001
Usher syndrome, type 1D	CDH23	AR	General Population	1 in 202	99%	1 in 20101
Usher syndrome, type 1F	PCDH15	AR	General Population	1 in 395	99%	1 in 39401
Usher syndrome, type 1F	PCDH15	AR	Ashkenazi Jewish	1 in 78	99%	1 in 7701
Usher syndrome, type 1J	CIB2	AR	General Population	<1 in 500	99%	<1 in 49901
Usher syndrome, type 2A	USH2A	AR	French Canadian	1 in 207	99%	1 in 20601
Usher syndrome, type 2A	USH2A	AR	General Population	1 in 126	99%	1 in 12501
Usher syndrome, type 2A	USH2A	AR	East Asian	1 in 113	99%	1 in 11201

Disease Name	Gene	Inheritance	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Usher syndrome, type 2A	USH2A	AR	Northern European Caucasian	1 in 113	99%	1 in 11201
Usher syndrome, type 2A	USH2A	AR	Caucasian	1 in 73	99%	1 in 7201
Usher syndrome, type 2A	USH2A	AR	Sephardic Jewish - Iraqi, Iranian	1 in 36	99%	1 in 3501
Usher syndrome, type 2C	ADGRV1	AR	General Population	1 in 176	99%	1 in 17501
Usher syndrome, type 3A	CLRN1	AR	General Population	<1 in 500	99%	<1 in 49901
Usher syndrome, type 3A	CLRN1	AR	Ashkenazi Jewish	1 in 120	99%	1 in 11901
Usher syndrome, type 3A	CLRN1	AR	Finnish	1 in 70	99%	1 in 6901
Very long-chain acyl-CoA dehydrogenase deficiency	ACADVL	AR	Asian	1 in 194	99%	1 in 19301
Very long-chain acyl-CoA dehydrogenase deficiency	ACADVL	AR	Caucasian	1 in 88	99%	1 in 8701
Very long-chain acyl-CoA dehydrogenase deficiency	ACADVL	AR	General Population	1 in 86	99%	1 in 8501
Vitamin D-dependent rickets, type 1A	CYP27B1	AR	General Population	1 in 22	99%	1 in 2101
Werner syndrome	WRN	AR	General Population	1 in 224	99%	1 in 22301
Wilson disease	ATP7B	AR	Southern European Caucasian	1 in 250	99%	1 in 24901
Wilson disease	ATP7B	AR	Northern European Caucasian	1 in 90	99%	1 in 8901
Wilson disease	ATP7B	AR	General Population	1 in 90	99%	1 in 8901
Wilson disease	ATP7B	AR	Caucasian	1 in 90	99%	1 in 8901
Wilson disease	ATP7B	AR	Ashkenazi Jewish	1 in 70	99%	1 in 6901
Wilson disease	ATP7B	AR	Sephardic Jewish - North African, Iraqi, Yemenite, Iranian, Bukharian	1 in 65	99%	1 in 6401
Wilson disease	ATP7B	AR	Sardinian	1 in 42	99%	1 in 4101
Wilson disease	ATP7B	AR	East Asian	1 in 27	99%	1 in 2601
Wilson disease	ATP7B	AR	Canary Islands	1 in 25	99%	1 in 2401
Wiskott-Aldrich syndrome, X-linked	WAS	XL	General Population	1 in 187500	99%	1 in 18749901
Wolcott-Rallison syndrome	EIF2AK3	AR	General Population	<1 in 500	99%	<1 in 49901
WWOX deficiency	WWOX	AR	General Population	<1 in 500	99%	<1 in 49901
Xeroderma pigmentosum, group A	XPA	AR	General Population	<1 in 500	99%	<1 in 49901
Xeroderma pigmentosum, group C	XPC	AR	General Population	<1 in 500	99%	<1 in 49901
X-linked agammaglobulinemia	BTK	XL	General Population	1 in 250000	99%	1 in 24999901
X-linked chondrodysplasia punctata 1	ARSL	XL	General Population	1 in 375000	99%	1 in 37499901
X-linked congenital adrenal hypoplasia	NR0B1	XL	General Population	1 in 52500	99%	1 in 5249901
X-linked developmental disorders, ARX-related	ARX	XL	General Population	<1 in 750000	50%	<1 in 1499999
X-linked heterotaxy-1	ZIC3	XL	General Population	<1 in 750000	99%	<1 in 74999901
X-Linked hyper IgM syndrome	CD40LG	XL	General Population	1 in 500000	99%	1 in 49999901
X-linked infantile spinal muscular atrophy	UBA1	XL	General Population	<1 in 750000	99%	<1 in 74999901
X-linked myotubular myopathy	MTM1	XL	General Population	1 in 38000	99%	1 in 3799901
X-linked Opitz G/BBB syndrome	MID1	XL	General Population	1 in 37500	99%	1 in 3749901
Zellweger spectrum disorders, PEX10-related	PEX10	AR	Asian	<1 in 500	99%	<1 in 49901
Zellweger spectrum disorders, PEX10-related	PEX10	AR	General Population	<1 in 500	99%	<1 in 49901
Zellweger spectrum disorders, PEX12-related	PEX12	AR	General Population	1 in 406	99%	1 in 40501
Zellweger spectrum disorders, PEX1-related	PEX1	AR	General Population	<1 in 500	99%	<1 in 49901
Zellweger spectrum disorders, PEX1-related	PEX1	AR	Caucasian	1 in 147	99%	1 in 14601
Zellweger spectrum disorders, PEX26-related	PEX26	AR	General Population	<1 in 500	99%	<1 in 49901
Zellweger spectrum disorders, PEX2-related	PEX2	AR	General Population	<1 in 500	99%	<1 in 49901
Zellweger spectrum disorders, PEX2-related	PEX2	AR	Caucasian	<1 in 500	99%	<1 in 49901
Zellweger spectrum disorders, PEX2-related	PEX2	AR	Ashkenazi Jewish	1 in 227	99%	1 in 22601
Zellweger spectrum disorders, PEX6-related	PEX6	AR	General Population	1 in 280	99%	1 in 27901
Zellweger spectrum disorders, PEX6-related	PEX6	AR	French Canadian	1 in 55	99%	1 in 5401
Zellweger spectrum disorders, PEX6-related	PEX6	AR	Sephardic Jewish - Yemenite	1 in 18	99%	1 in 1701