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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
ABCB11	Progressive familial intrahepatic cholestasis, type II	General Population	1 in 158	99%	1 in 15701
ABCC8	Familial hyperinsulinism, ABCC8-related	General Population	1 in 112	99%	1 in 11101
		Ashkenazi Jewish	1 in 52	99%	1 in 5101
		Finnish	1 in 29	99%	1 in 2801
ABCD1	Adrenoleukodystrophy, X-linked	General Population	1 in 10500	99%	1 in 1049901
		Sephardic Jewish	1 in 10500	99%	1 in 1049901
ACAD9	Riboflavin responsive complex 1 deficiency (acyl-coenzyme dehydrogenase 9 deficiency)	General Population	<1 in 500	99%	1 in 49901
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	General Population	1 in 35	99%	1 in 3401
		Asian	1 in 178	99%	1 in 17701
		Caucasian	1 in 64	99%	1 in 6300
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency	General Population	1 in 86	99%	1 in 8500
		Asian	1 in 194	99%	1 in 19301
		Caucasian	1 in 88	99%	1 in 8700
ACAT1	Beta-ketothiolase deficiency	General Population	1 in 347	99%	1 in 34601
		Asian	1 in 289	99%	1 in 28801
		Caucasian	1 in 354	99%	1 in 35301
ACOX1	Acyl-CoA oxidase I deficiency	General Population	<1 in 500	99%	1 in 49901
ACSF3	Combined malonic and methylmalonic aciduria	General Population	1 in 86	99%	1 in 8500
ADA	Adenosine deaminase deficiency	General Population	1 in 224	99%	1 in 22301
ADAMTS2	Ehlers-Danlos syndrome, type VIIC	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 248	99%	1 in 24701

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
ADGRG1	Bilateral frontoparietal polymicrogyria	General Population	<1 in 500	99%	1 in 49901
AGA	Aspartylglucosaminuria	General Population	<1 in 500	99%	1 in 49901
		Caucasian	<1 in 500	99%	1 in 49901
		Finnish	1 in 36	99%	1 in 3501
AGL	Glycogen storage disease, type III	General Population	1 in 159	99%	1 in 15801
		Faroese	1 in 28	99%	1 in 2701
		Sephardic Jewish - Moroccan	1 in 37	99%	1 in 3601
AGPS	Rhizomelic chondrodysplasia punctata, type III	General Population	<1 in 500	99%	1 in 49901
AGXT	Primary hyperoxaluria, type I	General Population	1 in 158	99%	1 in 15701
AIRE	Autoimmune polyglandular syndrome, type 1	General Population	1 in 354	99%	1 in 35301
		Finnish	1 in 79	99%	1 in 7800
		Sardinian	1 in 60	99%	1 in 5900
		Sephardic Jewish - Iranian	1 in 27	99%	1 in 2601
ALDH3A2	Sjogren-Larsson syndrome	General Population	1 in 223	99%	1 in 22201
		Swedish	1 in 204	99%	1 in 20301
ALDOB	Hereditary fructose intolerance	General Population	1 in 55	99%	1 in 5401
		African American	1 in 406	99%	1 in 40501
		Caucasian	1 in 80	99%	1 in 7900
		Hispanic	1 in 275	99%	1 in 27401
ALG6	Congenital disorder of glycosylation, type IC	General Population	<1 in 500	99%	1 in 49901
ALMS1	Alstrom syndrome	General Population	<1 in 500	99%	1 in 49901
ALPL	Hypophosphatasia	General Population	1 in 158	99%	1 in 15701
		Asian	1 in 203	99%	1 in 20201
		Mennonite	1 in 25	99%	1 in 2401

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
AMT	Glycine encephalopathy, AMT-related	General Population	1 in 262	99%	1 in 26101
		Caucasian	1 in 271	99%	1 in 27001
AQP2	Nephrogenic diabetes insipidus	General Population	<1 in 500	99%	1 in 49901
ARSA	Metachromatic leukodystrophy, ARSA-related	General Population	1 in 100	99%	1 in 9900
		Ashkenazi Jewish	<1 in 500	99%	1 in 49901
		Navajo	1 in 25	99%	1 in 2401
		Sephardic Jewish - Yemenite	1 in 46	99%	1 in 4501
ARSB	Mucopolysaccharidosis, type VI (Maroteaux-Lamy)	General Population	1 in 291	99%	1 in 29001
		Asian	1 in 423	99%	1 in 42201
		Caucasian	1 in 273	99%	1 in 27201
ASL	Argininosuccinate aciduria	General Population	1 in 133	99%	1 in 13201
ASNS	Asparagine synthetase deficiency	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Iranian	1 in 80	99%	1 in 7900
ASPA	Canavan disease	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 60	99%	1 in 5900
ASS1	Citrullinemia, type I	General Population	1 in 119	99%	1 in 11801
		Asian	1 in 123	99%	1 in 12201
		Caucasian	1 in 195	99%	1 in 19401
ATM	Ataxia-telangiectasia	General Population	1 in 100	99%	1 in 9900
		Ashkenazi Jewish	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Moroccan	1 in 81	99%	1 in 8000
ATP6V1B1	Renal tubular acidosis and deafness, ATP6V1B1-related	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Syrian	1 in 140	99%	1 in 13901

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ATP7A	Occipital horn syndrome (motor neuropathy, distal), X-linked	General Population	1 in 75000	99%	1 in 7499900
ATP7B	Wilson disease	General Population	1 in 90	99%	1 in 8900
		Ashkenazi Jewish	1 in 70	99%	1 in 6900
		Canary Islands	1 in 25	99%	1 in 2401
		Caucasian	1 in 90	99%	1 in 8900
		East Asian	1 in 27	99%	1 in 2601
		Sardinian	1 in 42	99%	1 in 4101
		Sephardic Jewish - North African, Iraqi, Yemenite, Iranian, Bukharian	1 in 65	99%	1 in 6400
ATRX	Alpha-thalassemia intellectual disability syndrome, X-linked	General Population	<1 in 750000	99%	1 in 74999900
BBS1	Bardet-Biedl syndrome, BBS1-related	General Population	1 in 265	99%	1 in 26401
		Faroese	1 in 30	99%	1 in 2901
BBS10	Bardet-Biedl syndrome, BBS10-related	General Population	1 in 447	99%	1 in 44601
BBS12	Bardet-Biedl syndrome, BBS12-related	General Population	<1 in 500	99%	1 in 49901
BBS2	Bardet-Biedl syndrome, BBS2-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 135	99%	1 in 13401
		Hutterites	1 in 22	99%	1 in 2101
BCKDHA	Maple syrup urine disease, type 1A	General Population	1 in 321	99%	1 in 32001
		Caucasian	1 in 320	99%	1 in 31901
		Mennonite	1 in 10	99%	1 in 900
		Portuguese Roma	1 in 71	99%	1 in 7000
BCKDHB	Maple syrup urine disease, type 1B	General Population	1 in 364	99%	1 in 36301
		Ashkenazi Jewish	1 in 97	99%	1 in 9600

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
BCKDHB	Maple syrup urine disease, type 1B	Asian	1 in 163	99%	1 in 16201
		Caucasian	1 in 433	99%	1 in 43201
BCS1L	GRACILE syndrome	General Population	1 in 111	99%	1 in 11001
		Caucasian	1 in 407	99%	1 in 40601
		Finnish	1 in 108	99%	1 in 10701
BLM	Bloom syndrome	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 140	99%	1 in 13901
BSND	Bartter syndrome, type IV	General Population	<1 in 500	99%	1 in 49901
BTD	Biotinidase deficiency	General Population	1 in 120	99%	1 in 11901
		Caucasian	1 in 12	99%	1 in 1101
		Hispanic	1 in 30	99%	1 in 2901
BTK	Agammaglobulinemia, X-linked	General Population	1 in 250000	99%	1 in 24999901
CAPN3	Limb-girdle muscular dystrophy, type 2A	General Population	1 in 602	99%	1 in 60100
		Amish	1 in 50	99%	1 in 4901
		Caucasian	1 in 103	99%	1 in 10201
		East Asian	1 in 232	99%	1 in 23101
		Hispanic	1 in 260	99%	1 in 25901
CBS	Homocystinuria, CBS-related	General Population	1 in 224	99%	1 in 22301
		Caucasian	1 in 52	99%	1 in 5101
		Qatari	1 in 21	99%	1 in 2001
CDH23	Usher syndrome, type ID	General Population	1 in 202	99%	1 in 20101
CEP290	Leber congenital amaurosis, CEP290-related	General Population	1 in 185	99%	1 in 18401

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
CERKL	Retinitis pigmentosa 26	General Population	1 in 137	99%	1 in 13601
		Sephardic Jewish - Yemenite	1 in 24	99%	1 in 2301
CFTR	Cystic fibrosis	General Population	1 in 25	99%	1 in 2401
		African American	1 in 61	99%	1 in 6000
		Ashkenazi Jewish	1 in 25	99%	1 in 2401
		Caucasian	1 in 25	99%	1 in 2401
		East Asian	1 in 90	99%	1 in 8900
		Hispanic	1 in 46	99%	1 in 4501
CHAT	Congenital myasthenic syndrome, CHAT-related	General Population	<1 in 500	99%	1 in 49901
CHM	Choroideremia, X-linked	General Population	1 in 25000	99%	1 in 2499901
CHRNE	Congenital myasthenic syndrome, CHRNE-related	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 383	99%	1 in 38201
CIITA	Bare lymphocyte syndrome, type II	General Population	<1 in 500	99%	1 in 49901
CLN3	Neuronal ceroid lipofuscinosis, CLN3-related	General Population	1 in 145	99%	1 in 14401
		Caucasian	1 in 188	99%	1 in 18701
CLN5	Neuronal ceroid lipofuscinosis, CLN5-related	General Population	1 in 317	99%	1 in 31601
		Finnish	1 in 289	99%	1 in 28801
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	General Population	1 in 261	99%	1 in 26001
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	General Population	1 in 349	99%	1 in 34801
		Finnish	1 in 135	99%	1 in 13401
CLRN1	Usher syndrome, type III	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 120	99%	1 in 11901
		Finnish	1 in 70	99%	1 in 6900

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CNGB3	Achromatopsia, CNGB3-related	General Population	1 in 98	99%	1 in 9700
		Caucasian	1 in 91	99%	1 in 9000
COL4A3	Alport syndrome, COL4A3-related	General Population	1 in 323	99%	1 in 32201
		Ashkenazi Jewish	1 in 189	99%	1 in 18801
		Caucasian	1 in 284	99%	1 in 28301
COL4A4	Alport syndrome, COL4A4-related	General Population	1 in 353	99%	1 in 35201
COL4A5	Alport syndrome, COL4A5-related, X-linked	General Population	1 in 47000	99%	1 in 4699901
COL7A1	Dystrophic epidermolysis bullosa, COL7A1-related	General Population	1 in 370	99%	1 in 36901
CPS1	Carbamoyl phosphate synthetase I deficiency	General Population	<1 in 500	99%	1 in 49901
		Asian	1 in 447	99%	1 in 44601
		Caucasian	1 in 284	99%	1 in 28301
CPT1A	Carnitine palmitoyl transferase I deficiency	General Population	<1 in 500	99%	1 in 49901
		Hutterites	1 in 16	99%	1 in 1501
CPT2	Carnitine palmitoyl transferase II deficiency	General Population	<1 in 500	99%	1 in 49901
		African American	1 in 308	99%	1 in 30701
		Ashkenazi Jewish	1 in 51	99%	1 in 5001
		Asian	<1 in 500	99%	1 in 49901
		Caucasian	1 in 200	99%	1 in 19901
CRB1	CRB1-related retinal dystrophies	General Population	1 in 112	99%	1 in 11101
CTNS	Cystinosis	General Population	1 in 158	99%	1 in 15701
		African American	<1 in 500	99%	1 in 49901
		Asian	<1 in 500	99%	1 in 49901
		Caucasian	1 in 220	99%	1 in 21901

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
CTNS	Cystinosis	French Canadian - Saguenay Lac-St. Jean	1 in 39	99%	1 in 3801
		Hispanic	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Moroccan	1 in 100	99%	1 in 9900
CTSK	Pycnodysostosis	General Population	1 in 439	99%	1 in 43801
CYBA	Chronic granulomatous disease, cytochrome b-negative	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Moroccan	1 in 13	99%	1 in 1201
CYBB	Chronic granulomatosis, X-linked	General Population	1 in 180000	99%	1 in 17999901
CYP11B2	Corticosterone methyloxidase deficiency	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Iranian	1 in 30	99%	1 in 2901
CYP17A1	Congenital adrenal hyperplasia, 17-alpha-hydroxylase deficiency	General Population	<1 in 500	99%	1 in 49901
CYP19A1	Aromatase deficiency	General Population	<1 in 500	99%	1 in 49901
CYP1B1	Primary congenital glaucoma	General Population	1 in 74	99%	1 in 7300
CYP21A2	Congenital adrenal hyperplasia, 21-hydroxylase-deficiency (non-classic)	General Population	1 in 17	99%	1 in 1601
		Ashkenazi Jewish	1 in 6.7	99%	1 in 571
		Caucasian	1 IN 10.5	99%	1 in 950
CYP21A2	Congenital adrenal hyperplasia, 21-hydroxylase-deficiency (classic)	General Population	1 in 61	99%	1 in 6000
CYP27A1	Cerebrotendinous xanthomatosis	General Population	1 in 115	99%	1 in 11401
		Sephardic Jewish - Moroccan	1 in 5	99%	1 in 401
DBT	Maple syrup urine disease, type 2	General Population	1 in 321	99%	1 in 32001
DCLRE1C	Omenn syndrome	General Population	<1 in 500	99%	1 in 49901
		Navajo and Apache Native Americans	1 in 10	99%	1 in 900

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DHCR7	Smith-Lemli-Opitz syndrome	General Population	1 in 100	99%	1 in 9900
		African American	1 in 93	99%	1 in 9200
		Ashkenazi Jewish	1 in 36	99%	1 in 3501
		Asian	<1 in 500	99%	1 in 49901
		Caucasian	1 in 50	99%	1 in 4901
DHDDS	Retinitis pigmentosa 59	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 333	99%	1 in 11601
DLD	Dihydrolipoamide dehydrogenase deficiency	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 94	99%	1 in 9300
DMD	Duchenne/Becker muscular dystrophy, X-linked	General Population	1 in 4200	99%	1 in 419901
DNAH5	Primary ciliary dyskinesia, DNAH5-related	General Population	1 in 120	99%	1 in 11901
		Ashkenazi Jewish	1 in 174	99%	1 in 17301
DNAI1	Primary ciliary dyskinesia, DNAI1-related	General Population	1 in 182	99%	1 in 18101
		Ashkenazi Jewish	1 in 352	99%	1 in 35101
DNAI2	Primary ciliary dyskinesia, DNAI2-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 200	99%	1 in 19901
DOK7	Congenital myasthenic syndrome, DOK7-related	General Population	1 in 454	99%	1 in 45301
		Caucasian	1 in 290	99%	1 in 28901
		French Canadian	1 in 353	99%	1 in 35201
DPYD	Dihydropyrimidine dehydrogenase deficiency	General Population	1 in 20	99%	1 in 1901
		East Asian	1 in 50	99%	1 in 4901
DYSF	Limb-girdle muscular dystrophy, type 2B	General Population	1 in 311	99%	1 in 31001
		Sephardic Jewish, Libyan, Moroccan, Tunisian, Bulgarian	1 in 14	99%	1 in 1301

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EDA	Hypohidrotic ectodermal dysplasia, X-linked	General Population	1 in 3800	99%	1 in 379901
EIF2B5	Leukoencephalopathy with vanishing white matter	General Population	<1 in 500	99%	1 in 49901
ELP1	Familial dysautonomia	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 34	99%	1 in 3301
EMD	Emery-Dreifuss muscular dystrophy, X-linked	General Population	1 in 375000	99%	1 in 37499901
ESCO2	Roberts syndrome	General Population	<1 in 500	99%	1 in 49901
ETFA	Glutaric acidemia, type IIA	General Population	<1 in 500	99%	1 in 49901
ETFDH	Glutaric acidemia, type IIC	General Population	1 in 250	99%	1 in 24901
		Asian	1 in 87	99%	1 in 8600
ETHE1	Ethylmalonic encephalopathy	General Population	<1 in 500	99%	1 in 49901
EVC	Ellis-van Creveld syndrome	General Population	1 in 345	99%	1 in 34401
		Lancaster County Amish	1 in 12	99%	1 in 1101
EYS	Retinitis pigmentosa 25	General Population	1 in 129	99%	1 in 12801
		Ashkenazi Jewish	<1 in 500	99%	1 in 49901
		Caucasian	1 in 53	99%	1 in 5201
		Sephardic Jewish - Moroccan	1 in 42	99%	1 in 4101
F11	Factor XI deficiency	General Population	1 in 92	99%	1 in 9100
		Ashkenazi Jewish	1 in 11	99%	1 in 1001
		Asian	1 in 163	99%	1 in 16201
		Caucasian	1 in 101	99%	1 in 10001
F9	Hemophilia B, X-linked	General Population	1 in 23000	99%	1 in 2299901
FAH	Tyrosinemia, type I	General Population	1 in 100	99%	1 in 9900
		African American	1 in 478	99%	1 in 47701

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FAH	Tyrosinemia, type I	Ashkenazi Jewish	1 in 150	99%	1 in 14901
		Asian	<1 in 500	99%	1 in 49901
		Caucasian	1 in 333	99%	1 in 33201
		French Canadian - Saguenay Lac-St. Jean	1 in 25	99%	1 in 2401
		French Canadian (Quebec)	1 in 66	99%	1 in 6501
FAM161A	Retinitis pigmentosa 28	General Population	1 in 289	99%	1 in 28801
		Ashkenazi Jewish	1 in 214	99%	1 in 21301
		Sephardic Jewish, Libyan, Moroccan, Tunisian, Bulgarian	1 in 41	99%	1 in 4001
FANCA	Fanconi anemia, type A	General Population	1 in 345	99%	1 in 34401
		Sephardic Jewish - Moroccan, Tunisian	1 in 133	99%	1 in 13201
		Spanish Roma	1 in 64	99%	1 in 6300
FANCC	Fanconi anemia, type C	General Population	1 in 1053	99%	1 in 105201
		Ashkenazi Jewish	1 in 98	99%	1 in 9700
FANCG	Fanconi anemia, type G	General Population	<1 in 500	99%	1 in 49901
		African American	1 in 100	99%	1 in 9900
FH	Fumarase deficiency	General Population	<1 in 500	99%	1 in 49901
FKRP	Limb-girdle muscular dystrophy, type 2I	General Population	1 in 158	99%	1 in 15701
		Norwegian	1 in 116	99%	1 in 11501
FKTN	Walker-Warburg syndrome, FKTN-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 150	99%	1 in 14901
		Japanese	1 in 188	99%	1 in 18701

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FMR1	Fragile X syndrome	General Population	1 in 250	99%	1 in 24901
		African American	1 in 251	99%	1 in 25001
		Ashkenazi Jewish	1 in 58	99%	1 in 5700
		Asian	<1 in 500	99%	1 in 49901
		Caucasian	1 in 178	99%	1 in 17701
		Hispanic	<1 in 500	99%	1 in 49901
G6PC	Glycogen storage disease, type Ia	General Population	1 in 177	99%	1 in 17601
		Ashkenazi Jewish	1 in 71	99%	1 in 7000
		Asian	1 in 192	99%	1 in 19101
		Caucasian	1 in 177	99%	1 in 17601
G6PD	Glucose-6-phosphate dehydrogenase deficiency, X-linked	General Population	1 in 30	99%	1 in 2901
		African American	1 in 5	99%	1 in 401
GAA	Glycogen storage disease, type II	General Population	1 in 132	99%	1 in 13101
		African American	1 in 70	99%	1 in 6900
		Ashkenazi Jewish	1 in 58	99%	1 in 5700
		Asian	1 in 112	99%	1 in 11101
		Caucasian	1 in 100	99%	1 in 9900
GALC	Krabbe disease	General Population	1 in 150	99%	1 in 14901
		Asian	<1 in 500	99%	1 in 49901
		Druze Northern Israel	1 in 6	99%	1 in 500
		Muslim Arab (Jerusalem)	1 in 6	99%	1 in 500
GALK1	Galactokinase deficiency	General Population	1 in 122	99%	1 in 12101
		Asian	<1 in 500	99%	1 in 49901
		Roma	1 in 47	99%	1 in 4601

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GALT	Galactosemia	General Population	1 in 110	99%	1 in 10901
		African American	1 in 78	99%	1 in 7700
		Ashkenazi Jewish	1 in 172	99%	1 in 17101
		Caucasian	1 in 108	99%	1 in 10701
		East Asian	<1 in 500	99%	1 in 49901
		Hispanic	1 in 305	99%	1 in 30401
		Irish Travellers	1 in 11	99%	1 in 1001
GAMT	Guanidinoacetate methyltransferase deficiency	General Population	<1 in 500	99%	1 in 49901
		Portuguese	1 in 125	99%	1 in 12401
GBA	Gaucher disease	General Population	1 in 153	99%	1 in 15201
		Ashkenazi Jewish	1 in 18	99%	1 in 1701
		Caucasian	1 in 164	99%	1 in 16301
GBE1	Glycogen storage disease, type IV	General Population	1 in 387	99%	1 in 38601
		Ashkenazi Jewish	1 in 68	99%	1 in 6701
		Caucasian	1 in 144	99%	1 in 14301
GCDH	Glutaric acidemia, type I	General Population	1 in 112	99%	1 in 11101
		African American	1 in 36	99%	1 in 3501
		Caucasian	1 in 172	99%	1 in 17101
		Lancaster County Amish	1 in 9	99%	1 in 1001
		Lumbee Native Americans	1 in 16	99%	1 in 1501
		Oji-Cree First Nations (N. Manitoba)	1 in 8	99%	1 in 700
GFM1	Combined oxidative phosphorylation deficiency 4	General Population	<1 in 500	99%	1 in 49901
GJB1	Charcot-Marie-Tooth disease, GJB1-related, X-linked	General Population	1 in 3700	99%	1 in 369901

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
GJB2	Nonsyndromic hearing loss, GJB2-related	General Population	1 in 42	99%	1 in 4101
		Ashkenazi Jewish	1 in 21	99%	1 in 2001
		Caucasian	1 in 30	99%	1 in 2901
		East Asian	1 in 40	99%	1 in 3901
GLA	Fabry disease, X-linked	General Population	1 in 42000	99%	1 in 4199901
GLB1	GM1 gangliosidosis	General Population	1 in 158	99%	1 in 15701
		Caucasian	1 in 278	99%	1 in 27701
		Maltese	1 in 30	99%	1 in 2901
		Roma	1 in 50	99%	1 in 4901
		South Brazil	1 in 65	99%	1 in 6400
GLDC	Glycine encephalopathy, GLDC-related	General Population	1 in 135	99%	1 in 13401
		Caucasian	1 in 140	99%	1 in 13901
GLE1	Lethal congenital contracture syndrome 1	General Population	<1 in 500	99%	1 in 49901
		Finnish	1 in 100	99%	1 in 9900
GNE	Inclusion body myopathy 2	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	<1 in 500	99%	1 in 49901
		Asian	1 in 58	99%	1 in 5700
		Caucasian	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Iranian, Syrian	1 in 12	99%	1 in 1101
GNPTAB	Mucolipidosis, type II/III alpha/beta	General Population	1 in 158	99%	1 in 15701
		Asian	1 in 389	99%	1 in 38801
		Caucasian	1 in 225	99%	1 in 22401
GNPTG	Mucolipidosis III gamma	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 In 273	99%	1 in 27201

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
GNS	Mucopolysaccharidosis, type IIID	General Population	<1 in 500	99%	1 in 49901
GRHPR	Primary hyperoxaluria, type II	General Population	<1 in 500	99%	1 in 49901
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	General Population	1 in 138	99%	1 in 13701
		Caucasian	1 in 254	99%	1 in 25301
		Finnish	1 in 240	99%	1 in 23901
HAX1	Congenital neutropenia, HAX1-related	General Population	<1 in 500	99%	1 in 49901
HBA1/HBA2	Alpha-thalassemia	General Population	1 in 25	99%	1 in 2401
		African American	1 in 30	99%	1 in 2901
		Asian	1 in 20	99%	1 in 1901
		Caucasian	<1 in 500	99%	1 in 49901
HBB	Beta-hemoglobinopathies	General Population	1 in 129	99%	1 in 12801
		African American	1 in 10	99%	1 in 900
		Asian	1 in 54	99%	1 in 5300
		Caucasian	1 in 373	99%	1 in 37201
		East Asian	1 in 78	99%	1 in 7700
		Hispanic	1 in 83	99%	1 in 8200
		Mediterranean	1 in 28	99%	1 in 2701
		Middle Eastern	1 in 5	99%	1 in 401
		South Asian	1 in 32	99%	1 in 3101
		Southeast Asian	1 in 30	99%	1 in 2901
HEXA	Tay-Sachs disease	General Population	1 in 250	99%	1 in 24901
		African American	1 in 271	99%	1 in 27001
		Ashkenazi Jewish	1 in 27	99%	1 in 2601

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
HEXA	Tay-Sachs disease	Asian	1 in 126	99%	1 in 12501
		Caucasian	1 in 182	99%	1 in 18101
		French Canadian - Gaspesie	1 in 13	99%	1 in 1201
		French Canadian - Other	1 in 73	99%	1 in 7200
		Irish	1 in 41	99%	1 in 4001
		Old Order Amish	1 in 3.4	99%	1 in 241
		Sephardic Jewish - Moroccan, Iraqi	1 in 125	99%	1 in 12401
HEXB	Sandhoff disease	General Population	1 in 278	99%	1 in 27701
		Argentinian Creole	1 in 64	99%	1 in 6300
		Ashkenazi Jewish	<1 in 500	99%	1 in 49901
		Caucasian	1 in 235	99%	1 in 23401
HGSNAT	Mucopolysaccharidosis, type IIIC (Sanfilippo C)	General Population	1 in 482	99%	1 in 48101
		Asian	<1 in 500	99%	1 in 49901
		Caucasian	1 in 259	99%	1 in 25801
HJV	Hereditary hemochromatosis, HJV-related	General Population	<1 in 500	99%	1 in 49901
		Caucasian	<1 in 500	99%	1 in 49901
HLCS	Holocarboxylase synthetase deficiency	General Population	<1 in 500	99%	1 in 49901
		Asian	1 in 158	99%	1 in 15701
		Caucasian	<1 in 500	99%	1 in 49901
		Faroese	1 in 20	99%	1 in 1901
HMGCL	3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	General Population	<1 in 500	99%	1 in 49901
HOGA1	Primary hyperoxaluria, type III	General Population	1 in 309	99%	1 in 30801

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
HPS1	Hermansky-Pudlak syndrome, HPS1-related	General Population	<1 in 500	99%	1 in 49901
		Puerto Rican	1 in 59	99%	1 in 5801
HPS3	Hermansky-Pudlak syndrome, HPS3-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 235	99%	1 in 23401
HSD17B4	D-Bifunctional protein deficiency	General Population	<1 in 158	99%	1 in 15701
HSD3B2	3-Beta-hydroxysteroid dehydrogenase deficiency, type II	General Population	<1 in 500	99%	1 in 49901
HYAL1	Mucopolysaccharidosis, type IX	General Population	<1 in 500	99%	1 in 49901
HYLS1	Hydrolethalus syndrome	General Population	1 in 455	99%	1 in 45401
		Finnish	1 in 50	99%	1 in 4901
IDS	Mucopolysaccharidosis, type II (Hunter syndrome)	General Population	1 in 75000	99%	1 in 7499900
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)	General Population	1 in 158	99%	1 in 15701
IL2RG	Severe combined immunodeficiency, IL2RG-related, X-linked	General Population	1 in 38000	99%	1 in 3799901
IVD	Isovaleric acidemia	General Population	1 in 250	99%	1 in 24901
		Asian	1 in 75	99%	1 in 7400
		Caucasian	1 in 144	99%	1 in 14301
KCNJ11	Familial hyperinsulinism, KCNJ11-related	General Population	<1 in 500	99%	1 in 49901
LAMA3	Junctional epidermolysis bullosa, LAMA3-related	General Population	<1 in 500	99%	1 in 49901
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	General Population	1 in 407	99%	1 in 40601
LAMC2	Junctional epidermolysis bullosa, LAMC2-related	General Population	1 in 500	99%	1 in 49901
LCA5	Leber congenital amaurosis, LCA5-related	General Population	<1 in 500	99%	1 in 49901
LDLRAP1	Familial hypercholesterolemia, LDLRAP1-related	General Population	<1 in 500	99%	1 in 49901
		Sardinian	1 in 143	99%	1 in 14201

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
LHX3	Combined pituitary hormone deficiency 3	General Population	<1 in 500	99%	1 in 49901
LIFR	Stuve-Wiedemann syndrome	General Population	<1 in 500	99%	1 in 49901
LIPA	Cholesteryl ester storage disease	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	<1 in 500	99%	1 in 49901
		Caucasian	1 in 145	99%	1 in 14401
		Sephardic Jewish - Iranian	1 in 26	99%	1 in 2501
LOXHD1	Deafness, autosomal recessive 77	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 180	99%	1 in 17901
LPL	Lipoprotein lipase deficiency	General Population	<1 in 500	99%	1 in 49901
		Asian	1 in 189	99%	1 in 18801
		Caucasian	<1 in 500	99%	1 in 49901
		French Canadian - Other	1 in 139	99%	1 in 13801
		French Canadian - Saguenay Lac-St. Jean	1 in 46	99%	1 in 4501
LRPPRC	Leigh syndrome, French-Canadian	General Population	<1 in 500	99%	1 in 49901
		French Canadian - Saguenay Lac-St. Jean	1 in 23	99%	1 in 2201
MAN2B1	Alpha-mannosidosis	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 485	99%	1 in 48401
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	General Population	1 in 147	99%	1 in 14601
		Caucasian	1 in 137	99%	1 in 13601
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	General Population	1 in 120	99%	1 in 11901
		Caucasian	1 in 112	99%	1 in 11101
MCOLN1	Mucolipidosis, type IV	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 89	99%	1 in 8800

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
MED17	Infantile cerebral and cerebellar atrophy	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Bukharian, Kurdish	1 in 20	99%	1 in 1901
MEFV	Familial Mediterranean fever	General Population	1 in 115	99%	1 in 11401
		Armenian	1 in 5	99%	1 in 401
		Ashkenazi Jewish	1 in 13	99%	1 in 1201
		Sephardic Jewish	1 in 14	99%	1 in 1301
		Turkish	1 in 5	99%	1 in 401
MESP2	Spondylothoracic dysostosis	General Population	1 in 224	99%	1 in 22301
		Puerto Rican	1 in 55	99%	1 in 5401
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	General Population	<1 in 500	99%	1 in 49901
MKS1	MKS1-related disorders	General Population	1 in 260	99%	1 in 25901
		Caucasian	1 in 260	99%	1 in 25901
		Finnish	1 in 47	99%	1 in 4601
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	General Population	<1 in 500	99%	1 in 49901
		Libyan Jewish	1 in 40	99%	1 in 3901
MMAA	Methylmalonic aciduria, MMAA-related	General Population	1 in 316	99%	1 in 31501
		Caucasian	1 in 316	99%	1 in 31501
MMAB	Methylmalonic aciduria, MMAB-related	General Population	1 in 456	99%	1 in 45501
		Caucasian	1 in 456	99%	1 in 45501
MMACHC	Methylmalonic aciduria, type cblC	General Population	1 in 138	99%	1 in 13701
		Asian	1 in 113	99%	1 in 11201
		Caucasian	1 in 138	99%	1 in 13701

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
MMADHC	Methylmalonic aciduria, type cblD	General Population	<1 in 500	99%	1 in 49901
		Caucasian	<1 in 500	99%	1 in 49901
MPI	Congenital disorder of glycosylation, type 1B	General Population	<1 in 500	99%	1 in 49901
		General Population	1 in 415	99%	1 in 41401
		Ashkenazi Jewish	1 in 57	99%	1 in 5600
		Caucasian	1 in 266	99%	1 in 26501
MPL	Congenital amegakaryocytic thrombocytopenia	General Population	1 in 415	99%	1 in 41401
		Ashkenazi Jewish	1 in 57	99%	1 in 5600
		Caucasian	1 in 266	99%	1 in 26501
MPV17	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	General Population	<1 in 500	99%	1 in 49901
		Navajo	1 in 20	99%	1 in 1901
MTM1	Myotubular myopathy, MTM1-related, X-linked	General Population	1 in 38000	99%	1 in 3799901
MTRR	Homocystinuria, type cblE	General Population	<1 in 500	99%	1 in 49901
		Caucasian	<1 in 500	99%	1 in 49901
MTTP	Abetalipoproteinemia	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 186	99%	1 in 18501
		Caucasian	<1 in 500	99%	1 in 49901
MUT	Methylmalonic aciduria, MUT-related	General Population	1 in 383	99%	1 in 38201
		African American	1 in 177	99%	1 in 17601
		Asian	1 in 53	99%	1 in 5201
		Caucasian	1 in 224	99%	1 in 22301
		Hispanic	1 in 383	99%	1 in 38201

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
MYO7A	Usher syndrome, type 1B	General Population	1 in 206	99%	1 in 20501
		African American	<1 in 500	99%	1 in 49901
MYO7A	Usher syndrome, type 1B	Asian	1 in 62	99%	1 in 6100
		Caucasian	1 in 145	99%	1 in 14401
NAGLU	Mucopolysaccharidosis, type IIIB (Sanfilippo B)	General Population	<1 in 500	99%	1 in 49901
		Asian	1 in 298	99%	1 in 29701
		Caucasian	1 in 346	99%	1 in 34501
NAGS	N-acetylglutamate synthase deficiency	General Population	<1 in 500	99%	1 in 49901
NBN	Nijmegen breakage syndrome	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 155	99%	1 in 15401
NDRG1	Charcot-Marie-Tooth disease, type 4D	General Population	<1 in 500	99%	1 in 49901
		Roma	1 in 22	99%	1 in 2101
NDUFAF5	Mitochondrial complex 1 deficiency, NDUFAF5-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 290	99%	1 in 28901
NDUFS6	Mitochondrial complex 1 deficiency, NDUFS6-related	General Population	<1 in 500	99%	1 in 49901
		Caucasus Jewish	1 in 24	99%	1 in 2301
NEB	Nemaline myopathy 2	General Population	1 in 224	99%	1 in 22301
		Ashkenazi Jewish	1 in 168	99%	1 in 16701
		Finnish	1 in 112	99%	1 in 11101
NPC1	Niemann-Pick disease, type CI/D	General Population	1 in 282	99%	1 in 28101
		Asian	1 in 404	99%	1 in 40301
		Caucasian	1 in 185	99%	1 in 18401
NPC2	Niemann-Pick disease, type CII	General Population	<1 in 500	99%	1 in 49901

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NPHP1	Juvenile nephronophthisis	General Population	1 in 202	99%	1 in 20101
NPHS1	Nephrotic syndrome, type 1	General Population	1 in 325	99%	1 in 32401
		Finnish	1 in 45	99%	1 in 4401
		Groffdale Conference Mennonites	1 in 12	99%	1 in 1101
NPHS2	Steroid resistant nephrotic syndrome	General Population	1 in 377	99%	1 in 37601
NR2E3	Enhanced S-Cone syndrome	General Population	1 in 204	99%	1 in 20301
		Ashkenazi Jewish	1 in 100	99%	1 in 9900
NTRK1	Congenital insensitivity to pain with anhidrosis (CIPA)	General Population	<1 in 500	99%	1 in 49901
		Asian	1 in 387	99%	1 in 38601
		Sephardic Jewish - Moroccan	<1 in 500	99%	1 in 49901
OAT	Ornithine aminotransferase deficiency	General Population	<1 in 500	99%	1 in 49901
		Finnish	1 in 147	99%	1 in 14601
		Sephardic Jewish - Iraqi, Syrian	1 in 177	99%	1 in 17601
OCRL	Lowe syndrome, X-linked	General Population	1 in 25000	99%	1 in 2499901
OPA3	3-Methylglutaconic aciduria, type III	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Iraqi	1 in 13	99%	1 in 1201
OTC	Ornithine transcarbamylase deficiency, X-linked	General Population	<1 in 30000	99%	1 in 2999901
PAH	Phenylalanine hydroxylase deficiency	General Population	1 in 65	99%	1 in 6400
		African American	1 in 143	99%	1 in 14201
		Ashkenazi Jewish	1 in 225	99%	1 in 22401
		Asian	1 in 78	99%	1 in 7700
		Caucasian	1 in 50	99%	1 in 4901
		Irish	1 in 34	99%	1 in 3301

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PAH	Phenylalanine hydroxylase deficiency	Sephardic Jewish - Iranian, Bukharian, Kavkazi, Tunisian, Moroccan	1 in 18	99%	1 in 1701
		Sicilian	1 in 26	99%	1 in 2501
		Turkish	1 in 32	99%	1 in 3101
PCCA	Propionic acidemia, PCCA-related	General Population	1 in 224	99%	1 in 22301
		Asian	1 in 162	99%	1 in 16101
		Caucasian	1 in 380	99%	1 in 37901
PCCB	Propionic acidemia, PCCB-related	General Population	1 in 224	99%	1 in 22301
		Asian	1 in 145	99%	1 in 14401
		Caucasian	1 in 202	99%	1 in 20101
PCDH15	Usher syndrome, type 1F	General Population	1 in 395	99%	1 in 39401
		Ashkenazi Jewish	1 in 78	99%	1 in 7700
PDHA1	Pyruvate dehydrogenase deficiency, PDHA1-related, X-linked	General Population	<1 in 750000	99%	1 in 74999900
PDHB	Pyruvate dehydrogenase deficiency, PDHB-related	General Population	<1 in 500	99%	1 in 49901
PEX1	Zellweger spectrum disorders, PEX1-related	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 147	99%	1 in 14601
PEX10	Zellweger spectrum disorders, PEX10-related	General Population	<1 in 500	99%	1 in 49901
		Asian	<1 in 500	99%	1 in 49901
PEX2	Zellweger spectrum disorders, PEX2-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 227	99%	1 in 22601
		Caucasian	<1 in 500	99%	1 in 49901
PEX6	Zellweger spectrum disorders, PEX6-related	General Population	1 in 280	99%	1 in 27901
		French Canadian	1 in 55	99%	1 in 5401
		Sephardic Jewish - Yemenite	1 in 18	99%	1 in 1701

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PEX7	Rhizomelic chondrodysplasia punctata, type I	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 158	99%	1 in 15701
PFKM	Glycogen storage disease, type VII	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 250	99%	1 in 24901
PHGDH	3-Phosphoglycerate dehydrogenase deficiency	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 453	99%	1 in 45201
PKHD1	Autosomal recessive polycystic kidney disease	General Population	1 in 144	99%	1 in 14301
		Ashkenazi Jewish	1 in 106	99%	1 in 10501
		Caucasian	1 in 100	99%	1 in 9900
		South African Afrikaner	1 in 52	99%	1 in 5101
PLA2G6	Infantile neuroaxonal dystrophy 1	General Population	<1 in 500	99%	1 in 49901
PMM2	Congenital disorder of glycosylation, type 1A	General Population	1 in 124	99%	1 in 12301
		Ashkenazi Jewish	1 in 61	99%	1 in 6000
		Asian	1 in 449	99%	1 in 44801
		Caucasian	1 in 42	99%	1 in 4101
POLG	POLG-related disorders	General Population	1 in 50	99%	1 in 4901
POMGNT1	Muscle-eye-brain disease, POMGNT1-related	General Population	1 in 462	99%	1 in 46101
		Finnish	1 in 111	99%	1 in 11001
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	General Population	1 in 368	99%	1 in 36701
		Finnish	1 in 70	99%	1 in 6900
PROP1	PROP1-related combined pituitary hormone deficiency	General Population	1 in 141	99%	1 in 14001
PSAP	Metachromatic leukodystrophy, PSAP-related	General Population	<1 in 500	99%	1 in 49901

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PTS	6-Pyruvoyl-tetrahydropterin synthase (PTPS) deficiency	General Population	<1 in 500	99%	1 in 49901
		Asian	1 in 122	99%	1 in 12101
PUS1	Mitochondrial myopathy and sideroblastic anemia	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Iranian	<1 in 500	99%	1 in 49901
PYGM	Glycogen storage disease, type V	General Population	1 in 191	99%	1 in 19001
		Caucasian	1 in 191	99%	1 in 19001
		Sephardic Jewish - Kurdish	1 in 84	99%	1 in 8300
RAB23	Carpenter syndrome	General Population	<1 in 500	99%	1 in 49901
		Caucasian	<1 in 500	99%	1 in 49901
RAG2	Omenn syndrome, RAG2-related	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Iraqi	<1 in 500	99%	1 in 49901
RAPSN	Congenital myasthenic syndrome, RAPSN-related	General Population	1 in 252	99%	1 in 25101
		Caucasian	1 in 176	99%	1 in 17501
		Sephardic Jewish - Iraqi, Iranian	<1 in 500	99%	1 in 49901
RARS2	Pontocerebellar hypoplasia, RARS2-related	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Iraqi, Syrian, Tunisian	<1 in 500	99%	1 in 49901
RDH12	Leber congenital amaurosis, RDH12-related	General Population	1 in 456	99%	1 in 45501
RMRP	Cartilage-hair hypoplasia	General Population	<1 in 500	99%	1 in 49901
		Amish	1 in 19	99%	1 in 1801
		Finnish	1 in 76	99%	1 in 7500
RPE65	Leber congenital amaurosis, RPE65-related	General Population	1 in 228	99%	1 in 22701
		Sephardic Jewish - North African	1 in 90	99%	1 in 8900
RPGRIP1L	Ciliopathies, RPGRIP1L-related	General Population	1 in 259	99%	1 in 25801

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
RS1	Juvenile retinoschisis, X-linked	General Population	1 in 2500	99%	1 in 249901
RTEL1	Dyskeratosis congenita, RTEL1-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 165	99%	1 in 16401
SACS	Spastic ataxia of Charlevoix-Saguenay (ARSACS)	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 450	99%	1 in 44901
		French Canadian - Charlevoix-Saguenay	1 in 21	99%	1 in 2001
SAMHD1	Aicardi-Goutières syndrome, SAMHD1-related	General Population	<1 in 500	99%	1 in 49901
SEPSECS	Progressive cerebello-cerebral atrophy	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Moroccan, Iraqi	1 in 41	99%	1 in 4001
SGCA	Limb-girdle muscular dystrophy, type 2D	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 290	99%	1 in 28901
		Finnish	1 in 150	99%	1 in 14901
SGCB	Limb-girdle muscular dystrophy, type 2E	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 406	99%	1 in 40501
SGCG	Limb-girdle muscular dystrophy, type 2C	General Population	1 in 354	99%	1 in 35301
		Moroccan	1 in 250	99%	1 in 24901
		Roma	1 in 96	99%	1 in 9500
SGSH	Mucopolysaccharidosis, type IIIA (Sanfilippo A)	General Population	1 in 415	99%	1 in 41401
		Caucasian	1 in 253	99%	1 in 25201
SLC12A3	Gitelman syndrome	General Population	1 in 100	99%	1 in 9900
SLC12A6	Andermann syndrome (hereditary motor and sensory neuropathy with agenesis of the corpus callosum)	General Population	<1 in 500	99%	1 in 49901
		French Canadian	1 in 23	99%	1 in 2201

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
SLC17A5	Salla disease	General Population	<1 in 500	99%	1 in 49901
		Canadian Inuit	1 in 129	99%	1 in 12801
		Finnish	1 in 100	99%	1 in 9900
		Swedish	1 in 125	99%	1 in 12401
SLC22A5	Carnitine deficiency, systemic primary	General Population	1 in 200	99%	1 in 19901
		Asian	1 in 100	99%	1 in 9900
		Caucasian	1 in 110	99%	1 in 10901
		Faroese	1 in 20	99%	1 in 1901
SLC25A13	Citrin deficiency	General Population	<1 in 500	99%	1 in 49901
		Asian	1 in 123	99%	1 in 12201
		Caucasian	<1 in 500	99%	1 in 49901
SLC25A15	Ornithine translocase deficiency	General Population	<1 in 500	99%	1 in 49901
		Metis from Saskatchewan	1 in 19	99%	1 in 1801
SLC26A2	SLC26A2-related skeletal dysplasias	General Population	1 in 158	99%	1 in 15701
		Finnish	1 in 50	99%	1 in 4901
SLC26A4	Pendred syndrome	General Population	1 in 80	99%	1 in 7900
		African American	1 in 76	99%	1 in 7500
		Asian	1 in 74	99%	1 in 7300
		Caucasian	1 in 88	99%	1 in 8700
SLC35A3	Arthrogryposis, mental retardation and seizures	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 453	99%	1 in 45201
SLC37A4	Glycogen storage disease, type Ib	General Population	1 in 354	99%	1 in 35301
		Caucasian	<1 in 500	99%	1 in 49901

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SLC39A4	Acrodermatitis enteropathica	General Population	1 in 354	99%	1 in 35301
SLC4A11	Corneal dystrophy and perceptive deafness syndrome	General Population	<1 in 500	99%	1 in 49901
SLC6A8	Creatine transporter defect, SLC6A8-related, X-linked	General Population	<1 in 410000	99%	1 in 40999901
SLC7A7	Lysinuric protein intolerance	General Population	<1 in 500	99%	1 in 49901
		Finnish	1 in 122	99%	1 in 12101
		Japanese	1 in 119	99%	1 in 11801
SMARCAL1	Schimke immunoosseous dysplasia	General Population	<1 in 500	99%	1 in 49901
SMN1	Spinal muscular atrophy	General Population	1 in 54	91%	1 in 589
		African American	1 in 72	71%	1 in 245
		Ashkenazi Jewish	1 in 67	99%	1 in 6600
		Asian	1 in 59	99%	1 in 5801
		Caucasian	1 in 47	95%	1 in 920
		Hispanic	1 in 117	90%	1 in 1161
SMPD1	Niemann-Pick disease, type A/B	General Population	1 in 196	99%	1 in 19501
		Ashkenazi Jewish	1 in 115	99%	1 in 11401
		Caucasian	1 in 244	99%	1 in 24301
STAR	Congenital lipoid adrenal hyperplasia	General Population	<1 in 500	99%	1 in 49901
		East Asian	1 in 177	99%	1 in 17601
SUMF1	Multiple sulfatase deficiency	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 279	99%	1 in 27801
TCIRG1	Osteopetrosis, infantile malignant, TCIRG1-related	General Population	1 in 316	99%	1 in 31501
		Ashkenazi Jewish	1 in 350	99%	1 in 34901
		Chuvashiya	1 in 60	99%	1 in 5900
		Costa Rican	1 in 86	99%	1 in 8500

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TECPR2	Hereditary spastic paraparesis, type 49	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Bukharian	1 in 27	99%	1 in 2601
TFR2	Hereditary hemochromatosis, TFR2-related	General Population	<1 in 500	99%	1 in 49901
TGM1	Lamellar ichthyosis, type 1	General Population	1 in 301	99%	1 in 30001
		Caucasian	1 in 253	99%	1 in 25201
		Norwegian	1 in 151	99%	1 in 15001
TH	Tyrosine hydroxylase deficiency	General Population	<1 in 500	99%	1 in 49901
		Asian	1 in 416	99%	1 in 41501
		Caucasian	1 in 224	99%	1 in 22301
TMEM216	Joubert syndrome 2	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 110	99%	1 in 10901
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	General Population	1 in 314	99%	1 in 31301
		Newfoundland	1 in 59	99%	1 in 5801
TRMU	Acute infantile liver failure	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Yemenite	1 in 34	99%	1 in 3301
TSFM	Combined oxidative phosphorylation deficiency 3	General Population	<1 in 500	99%	1 in 49901
		Finnish	1 in 80	99%	1 in 7900
TTPA	Ataxia with vitamin E deficiency	General Population	<1 in 500	99%	1 in 49901
		Caucasian	<1 in 500	99%	1 in 49901
TYMP	Myoneurogastrointestinal encephalopathy (MNGIE)	General Population	<1 in 500	99%	1 in 49901
		Caucasian	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Iranian	1 in 158	99%	1 in 15701

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USH1C	Usher syndrome, type 1C	General Population	1 in 353	99%	1 in 35201
		French Canadian / Acadian	1 in 227	99%	1 in 22601
USH2A	Usher syndrome, type IIA	General Population	1 in 126	99%	1 in 12501
		Caucasian	1 in 73	99%	1 in 7200
		Sephardic Jewish - Iraqi, Iranian	1 in 36	99%	1 in 3501
VPS13A	Choreoacanthocytosis	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	<1 in 500	99%	1 in 49901
VPS13B	Cohen syndrome	General Population	<1 in 500	99%	1 in 49901
VPS45	Congenital neutropenia, VPS45-related	General Population	<1 in 500	99%	1 in 49901
VRK1	Pontocerebellar hypoplasia, VRK1-related	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 225	99%	1 in 22401
VSX2	Microphthalmia/anophthalmia	General Population	<1 in 500	99%	1 in 49901
		Sephardic Jewish - Iranian, Syrian	1 in 145	99%	1 in 14401
WAS	Wiskott-Aldrich syndrome, X-linked	General Population	1 in 67000	99%	1 in 6699900
WNT10A	Odonto-onycho-dermal dysplasia/Schopf-Schulz-Passarge syndrome	General Population	1 in 305	99%	1 in 30401

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