Avero® Exon

150 genes

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
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
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
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
AGXT	Primary hyperoxaluria, type I	General Population	1 in 158	99%	1 in 15701

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
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
AMT	Glycine encephalopathy, AMT-related	General Population	1 in 262	99%	1 in 26101
		Caucasian	1 in 271	99%	1 in 27001
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
ASL	Argininosuccinate aciduria	General Population	1 in 133	99%	1 in 13201
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
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
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
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
		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
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
CAPN3	Limb-girdle muscular dystrophy, type 2A	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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
CDH23	Usher syndrome, type ID	General Population	1 in 202	99%	1 in 20101
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
CHRNE	Congenital myasthenic syndrome, CHRNE-related	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 383	99%	1 in 38201
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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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
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
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
		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
CYBB	Chronic granulomatosis, X-linked	General Population	1 in 180000	99%	1 in 17999901
CYP1B1	Primary congenital glaucoma	General Population	1 in 74	99%	1 in 7300

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
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
ETHE1	Ethylmalonic encephalopathy	General Population	<1 in 500	99%	1 in 49901
FAH	Tyrosinemia, type I	General Population	1 in 100	99%	1 in 9900
		African American	1 in 478	99%	1 in 47701
		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
FANCC	Fanconi anemia, type C	General Population	1 in 1053	99%	1 in 105201
		Ashkenazi Jewish	1 in 98	99%	1 in 9700
FH	Fumarase deficiency	General Population	<1 in 500	99%	1 in 49901
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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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
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
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
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
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
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
		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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
HEXA	Tay-Sachs disease	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
HMGCL	3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	General Population	<1 in 500	99%	1 in 49901
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
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
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
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
MAN2B1	Alpha-mannosidosis	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 485	99%	1 in 48401
MCOLN1	Mucolipidosis, type IV	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 89	99%	1 in 8800
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	General Population	<1 in 500	99%	1 in 49901
		Libyan Jewish	1 in 40	99%	1 in 3901
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
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
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

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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
		Asian	1 in 62	99%	1 in 6100
		Caucasian	1 in 145	99%	1 in 14401
NBN	Nijmegen breakage syndrome	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 155	99%	1 in 15401
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
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
OCRL	Lowe syndrome, X-linked	General Population	1 in 25000	99%	1 in 2499901
OTC	Ornithine transcarbamylase deficiency, X-linked	General Population	<1 in 30000	99%	1 in 2999901

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
		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
PC	Pyruvate carboxylase deficiency	General Population	1 in 250	99%	1 in 24901
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
PEX1	Zellweger spectrum disorders, PEX1-related	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 147	99%	1 in 14601

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
PEX7	Rhizomelic chondrodysplasia punctata, type I	General Population	<1 in 500	99%	1 in 49901
		Caucasian	1 in 158	99%	1 in 15701
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
PROP1	PROP1-related combined pituitary hormone deficiency	General Population	1 in 141	99%	1 in 14001
PTS	6-Pyruvoyl-tetrahydropterin synthase (PTPS) deficiency	General Population	<1 in 500	99%	1 in 49901
		Asian	1 in 122	99%	1 in 12101
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
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
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
SERPINA1	Alpha-1 antitrypsin deficiency	General Population	1 in 38	99%	1 in 3701
		Ashkenazi Jewish	1 in 24	99%	1 in 2301
		Northern European Caucasian	1 in 15	99%	1 in 1401
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
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
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
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
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
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

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GENE	DISORDER NAME	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RESIDUAL RISK
SUMF1	Multiple sulfatase deficiency	General Population	<1 in 500	99%	1 in 49901
		Ashkenazi Jewish	1 in 279	99%	1 in 27801
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
TTPA	Ataxia with vitamin E deficiency	General Population	<1 in 500	99%	1 in 49901
		Caucasian	<1 in 500	99%	1 in 49901
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
WAS	Wiskott-Aldrich syndrome, X-linked	General Population	1 in 67000	99%	1 in 6699900