Carrier screening gene list and carrier detection rates

This table displays residual risks after a negative result for each of the genes and corresponding disorders. The values provided assume a negative family history and the absence of symptoms for each disorder. Residual risk values are provided for disorders when carrier frequency is greater than 1 in 500. For disorders with carrier frequency equal to, or less than, 1 in 500, residual risk is considered to be reduced substantially. When provided, residual risk values are inferred from published carrier frequencies and estimated detection rates are based on testing technologies used at Invitae. Residual risks are provided only as a guide for assessing approximate risk given a negative result; values will vary based on the exact ethnic background of an individual. With individuals of mixed ethnicity, it is recommended to use the highest residual risk estimate. ***For any genes marked with an asterisk, an accurate residual risk value could not be calculated due to sample-specific limitations. Refer to the Limitations section of the patient report for detailed coverage information.***

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	DETECTION RATE	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
3-beta-hydroxysteroid dehydrogenase type II deficiency (congenital adrenal hyperplasia) (AR) NM_000198.3	HSD3B2	Pan-ethnic	≤1 in 500	99%	Reduced
3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency (AR) NM_000191.2	HMGCL	Pan-ethnic	≤1 in 500	99%	Reduced
		Portuguese	1 in 160	99%	1 in 15,900
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC1-related) (AR) NM_020166.4	MCCC1	Pan-ethnic	1 in 134	99%	1 in 13,300
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC2-related) (AR) NM_022132.4	MCCC2	Pan-ethnic	1 in 134	99%	1 in 13,300
3-methylglutaconic aciduria type III (Costeff optic atrophy) (AR) NM_025136.3	OPA3	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish (Iraqi)	1 in 10	99%	1 in 900
11-beta-hydroxylase-deficient congenital adrenal hyperplasia (AR) NM_000497.3	CYP11B1	Pan-ethnic	1 in 194	99%	1 in 19,300
		Sephardic Jewish (Moroccan)	1 in 40	99%	1 in 3,900
17-alpha-hydroxylase-deficient congenital adrenal hyperplasia (AR) NM_000102.3	CYP17A1	Pan-ethnic	≤1 in 500	99%	Reduced
Abetalipoproteinemia (AR) NM_000253.3	MTTP	Ashkenazi Jewish	1 in 131	99%	1 in 13,000
		Pan-ethnic	≤1 in 500	99%	Reduced
ACAD9 deficiency (AR) NM_014049.4	ACAD9	Pan-ethnic	≤1 in 500	99%	Reduced
Achromatopsia (AR) NM_019098.4	CNGB3	Pan-ethnic	1 in 93	99%	1 in 9,200
Acrodermatitis enteropathica (AR) NM_130849.3	SLC39A4	Pan-ethnic	1 in 354	99%	1 in 35,300
ADA-related conditions (AR) NM_000022.2	ADA	Pan-ethnic	1 in 224	92%	1 in 2,788
Aicardi-Goutières syndrome (AR) NM_015474.3	SAMHD1	Pan-ethnic	≤1 in 500	99%	Reduced
Aldosterone synthase deficiency (AR) NM_000498.3	CYP11B2	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish (Iranian)	1 in 30	99%	1 in 2,900
Alkaptonuria (AR) NM_000187.3	HGD	Pan-ethnic	1 in 250	99%	1 in 24,900
		Slovakian	1 in 69	99%	1 in 6,800
Alpha-1 antitrypsin deficiency (AR) NM_000295.4	SERPINA1	African-American	1 in 29	95%	1 in 560
		East Asian	1 in 249	95%	1 in 4,960
		Hispanic	1 in 9	95%	1 in 160
		Northern European	1 in 10	95%	1 in 180
		Pan-ethnic	1 in 13	95%	1 in 240

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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	DETECTION RATE	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Alpha-mannosidosis (AR) NM_000528.3	MAN2B1	Pan-ethnic	1 in 354	99%	1 in 35,300
Alpha-thalassemia (AR) NM_000558.4, NM_000517.4	HBA1/ HBA2*	African-American	1 in 30	90%	1 in 291
		Asian	1 in 20	90%	1 in 191
		Caucasian	≤1 in 500	90%	Reduced
		Pan-ethnic	1 in 25	90%	1 in 241
Alpha-thalassemia X-linked intellectual disability syndrome (XL) NM_000489.4	ATRX	Pan-ethnic	≤1 in 500	99%	Reduced
Alport syndrome (AR) NM_000091.4	COL4A3	Ashkenazi Jewish	1 in 192	99%	1 in 19,100
		Caucasian	1 in 284	99%	1 in 28,300
		Pan-ethnic	1 in 354	99%	1 in 35,300
Alport syndrome (AR) NM_000092.4	COL4A4	Pan-ethnic	1 in 353	99%	1 in 35,200
Alport syndrome, X-linked (XL) NM_000495.4	COL4A5*	Pan-ethnic	≤1 in 500	98%	Reduced
Alstrom syndrome (AR) NM_015120.4	ALMS1	Pan-ethnic	≤1 in 500	99%	Reduced
Andermann syndrome (AR) NM_133647.1	SLC12A6	French Canadian (Saguenay–Lac-St-Jean)	1 in 23	99%	1 in 2,200
		Pan-ethnic	≤1 in 500	99%	Reduced
Arginase deficiency (AR) NM_000045.3	ARG1	Pan-ethnic	1 in 274	99%	1 in 27,300
Argininosuccinic aciduria (AR) NM_000048.3	ASL	Pan-ethnic	1 in 133	90%	1 in 1,321
Aromatase deficiency (AR) NM_031226.2	CYP19A1	Pan-ethnic	≤1 in 500	99%	Reduced
Asparagine synthetase deficiency (AR) NM_133436.3	ASNS	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish (Iranian)	1 in 80	99%	1 in 7,900
Aspartylglucosaminuria (AR) NM_000027.3	AGA	Finnish	1 in 69	99%	1 in 6,800
		Pan-ethnic	≤1 in 500	99%	Reduced
Ataxia telangiectasia (AR) NM_000051.3	ATM	Pan-ethnic	1 in 100	99%	1 in 9,900
		Sephardic Jewish	1 in 69	99%	1 in 6,800
Ataxia with vitamin E deficiency (AR) NM_000370.3	TTPA	Italian	1 in 274	90%	1 in 2,731
		Pan-ethnic	≤1 in 500	90%	Reduced
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia (AR) NM_000383.3	AIRE	Finnish	1 in 79	99%	1 in 7,800
		Pan-ethnic	1 in 150	99%	1 in 14,900
		Sardinian	1 in 60	99%	1 in 5,900
		Sephardic Jewish (Iranian)	1 in 48	99%	1 in 4,700
Autosomal recessive deafness 77 (AR) NM_144612.6	LOXHD1	Ashkenazi Jewish	1 in 180	99%	1 in 17,900
		Pan-ethnic	≤1 in 500	99%	Reduced
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) (AR) NM_014363.5	SACS	French Canadian (Saguenay–Lac-St-Jean)	1 in 21	99%	1 in 2,000
		Pan-ethnic	≤1 in 500	99%	Reduced
Bardet-Biedl syndrome (AR) NM_024649.4	BBS1	Faroese	1 in 30	99%	1 in 2,900
		Pan-ethnic	1 in 330	99%	1 in 32,900

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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	DETECTION RATE	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Bardet-Biedl syndrome (AR) NM_031885.3	BBS2	Ashkenazi Jewish	1 in 140	99%	1 in 13,900
		Pan-ethnic	1 in 560	99%	Reduced
Bardet-Biedl syndrome (AR) NM_024685.3	BBS10	Pan-ethnic	1 in 354	99%	1 in 35,300
Bardet-Biedl syndrome (AR) NM_152618.2	BBS12	Pan-ethnic	1 in 708	99%	Reduced
Bartter syndrome type IV (BSND-related) (AR) NM_057176.2	BSND	Pan-ethnic	≤1 in 500	99%	Reduced
Bernard-Soulier syndrome (AR) NM_000173.6	GP1BA	Pan-ethnic	≤1 in 500	84%	Reduced
Bernard-Soulier syndrome (AR) NM_000174.4	GP9	Pan-ethnic	≤1 in 500	99%	Reduced
Beta-ketothiolase deficiency (AR) NM_000019.3	ACAT1	Caucasian	1 in 354	99%	1 in 35,300
		Pan-ethnic	≤1 in 500	99%	Reduced
Biotinidase deficiency (AR) NM_000060.3	BTD	Pan-ethnic	1 in 125	99%	1 in 12,400
Bloom syndrome (AR) NM_000057.3	BLM	Ashkenazi Jewish	1 in 100	99%	1 in 9,900
		Pan-ethnic	≤1 in 500	99%	Reduced
Canavan disease (AR) NM_000049.2	ASPA	Ashkenazi Jewish	1 in 57	99%	1 in 5,600
		Pan-ethnic	1 in 159	99%	1 in 15,800
Carbamoylphosphate synthetase I deficiency (AR) NM_001875.4	CPS1	Pan-ethnic	≤1 in 500	99%	Reduced
Carnitine palmitoyltransferase I deficiency (AR) NM_001876.3	CPT1A	Hutterite	1 in 16	99%	1 in 1,500
		Pan-ethnic	≤1 in 500	99%	Reduced
Carnitine palmitoyltransferase II deficiency (AR) NM_000098.2	CPT2	Ashkenazi Jewish	1 in 45	99%	1 in 4,400
		Pan-ethnic	1 in 182	99%	1 in 18,100
Carpenter syndrome (RAB23-related) (AR) NM_183227.2	RAB23	Pan-ethnic	≤1 in 500	99%	Reduced
Cartilage-hair hypoplasia – anauxetic dysplasia spectrum disorders (AR) NR_003051.3	RMRP	Amish	1 in 10	99%	1 in 900
		Finnish	1 in 76	99%	1 in 7,500
		Pan-ethnic	≤1 in 500	99%	Reduced
Cerebrotendinous xanthomatosis (AR) NM_000784.3	CYP27A1	Pan-ethnic	1 in 112	98%	1 in 5,500
		Sephardic Jewish	1 in 76	98%	1 in 3,750
Charcot-Marie-Tooth disease (AR) NM_006096.3	NDRG1	Roma	1 in 22	99%	1 in 2,100
Charcot-Marie-Tooth disease, X-linked (XL) NM_000166.5	GJB1	Pan-ethnic	≤1 in 500	99%	Reduced
Chorea-acanthocytosis (AR) NM_033305.2	VPS13A*	Pan-ethnic	≤1 in 500	97%	Reduced
Choroideremia (XL) NM_000390.2	CHM	Pan-ethnic	≤1 in 500	95%	Reduced
Chronic granulomatous disease (AR) NM_000101.3	CYBA	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish (Moroccan)	1 in 13	99%	1 in 1,200
Chronic granulomatous disease (XL) NM_000397.3	CYBB	Pan-ethnic	≤1 in 500	99%	Reduced

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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	DETECTION RATE	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Citrin deficiency (AR) NM_014251.2	SLC25A13	Chinese	1 in 65	99%	1 in 6,400
		Japanese	1 in 65	99%	1 in 6,400
		Korean	1 in 112	99%	1 in 11,100
		Southern Chinese and Taiwanese	1 in 48	99%	1 in 4,700
Citrullinemia type 1 (AR) NM_000050.4	ASS1	Pan-ethnic	1 in 120	96%	1 in 2,975
Cockayne syndrome type A (AR) NM_000082.3	ERCC8	Pan-ethnic	1 in 514	99%	Reduced
Cockayne syndrome type B (AR) NM_000124.3	ERCC6	Pan-ethnic	1 in 377	99%	1 in 37,600
Cohen syndrome (AR) NM_017890.4	VPS13B	Amish (Ohio)	1 in 12	99%	1 in 1,100
		Pan-ethnic	≤1 in 500	99%	Reduced
Combined malonic and methylmalonic aciduria (AR) NM_174917.4	ACSF3	Pan-ethnic	1 in 87	99%	1 in 8,600
Combined oxidative phosphorylation deficiency (AR) NM_024996.5	GFM1	Pan-ethnic	≤1 in 500	99%	Reduced
Combined oxidative phosphorylation deficiency (AR) NM_001172696.1	TSFM*	Finnish	1 in 80	93%	1 in 1,129
		Pan-ethnic	≤1 in 500	93%	Reduced
Combined pituitary hormone deficiency (AR) NM_014564.4	LHX3	Pan-ethnic	≤1 in 500	99%	Reduced
Combined pituitary hormone deficiency (AR) NM_006261.4	PROP1	Pan-ethnic	1 in 45	98%	1 in 2,200
Congenital amegakaryocytic thrombocytopenia (AR) NM_005373.2	MPL	Ashkenazi Jewish	1 in 57	99%	1 in 5,600
		Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation (AR) NM_013339.3	ALG6*	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation (AR) NM_002435.2	MPI	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation (PMM2-related) (AR) NM_000303.2	PMM2	Ashkenazi Jewish	1 in 61	99%	1 in 6,000
		Caucasian	1 in 60	99%	1 in 5,900
		Pan-ethnic	1 in 190	99%	1 in 18,900
Congenital ichthyosis (AR) NM_000359.2	TGM1	Norwegian	1 in 151	95%	1 in 3,000
		Pan-ethnic	1 in 224	95%	1 in 4,460
Congenital insensitivity to pain with anhidrosis (AR) NM_001012331.1	NTRK1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital myasthenic syndrome (AR) NM_000080.3	CHRNE	European Roma	1 in 25	99%	1 in 2,400
		Pan-ethnic	1 in 200	99%	1 in 19,900
Congenital myasthenic syndrome (AR) NM_005055.4	RAPSN	Pan-ethnic	1 in 283	99%	1 in 28,200
Congenital neutropenia (AR) NM_006118.3	HAX1	Pan-ethnic	≤1 in 500	99%	Reduced
Corneal dystrophy and perceptive deafness (AR) NM_032034.3	SLC4A11	Pan-ethnic	≤1 in 500	99%	Reduced

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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	DETECTION RATE	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Cystic fibrosis and other CFTR-related disorders (AR) NM_000492.3	CFTR	African-American - classic CF	1 in 61	99%	1 in 6,000
		Ashkenazi Jewish - classic CF	1 in 29	99%	1 in 2,800
		Asian - classic CF	1 in 88	99%	1 in 8,700
		Caucasian - classic CF	1 in 28	99%	1 in 2,700
		Pan-ethnic - classic CF	1 in 45	99%	1 in 4,400
		Pan-ethnic - classic CF + CFTR-related disorders	1 in 9	99%	1 in 800
Cystinosis (AR) NM_004937.2	CTNS	French Canadian (Saguenay–Lac-St-Jean)	1 in 39	99%	1 in 3,800
		Pan-ethnic	1 in 158	99%	1 in 15,700
		Sephardic Jewish (Moroccan)	1 in 100	99%	1 in 9,900
D-bifunctional protein deficiency (AR) NM_000414.3	HSD17B4	Pan-ethnic	1 in 158	99%	1 in 15,700
DHDDS-related disorders (AR) NM_024887.3	DHDDS	Ashkenazi Jewish	1 in 117	99%	1 in 11,600
Dihydrolipoamide dehydrogenase deficiency (AR) NM_000108.4	DLD	Ashkenazi Jewish	1 in 107	98%	1 in 5,300
		Pan-ethnic	≤1 in 500	99%	Reduced
DMD-related dystrophinopathy (XL) NM_004006.2	DMD	Pan-ethnic	1 in 667	99%	Reduced
Dystrophic epidermolysis bullosa (AR) NM_000094.3	COL7A1	Pan-ethnic	1 in 370	97%	1 in 12,300
Ehlers-Danlos syndrome, dermatosparaxis type (AR) NM_014244.4	ADAMTS2	Ashkenazi Jewish	1 in 187	99%	1 in 18,600
		Pan-ethnic	≤1 in 500	99%	Reduced
Ellis-Van Creveld syndrome (AR) NM_147127.4	EVC2	Pan-ethnic	1 in 199	99%	1 in 19,800
Ellis-Van Creveld syndrome (AR) NM_153717.2	EVC	Amish	1 in 8	99%	1 in 700
		Pan-ethnic	1 in 220	99%	1 in 21,900
Emery-Dreifuss muscular dystrophy (EMD-related) (AR) NM_000117.2	EMD	Pan-ethnic	≤1 in 500	99%	Reduced
Enhanced S-cone syndrome/retinitis pigmentosa 37 (AR) NM_014249.3	NR2E3	Pan-ethnic	≤1 in 500	99%	Reduced
Ethylmalonic encephalopathy (AR) NM_014297.3	ETHE1	Pan-ethnic	≤1 in 500	99%	Reduced
Fabry disease (XL) NM_000169.2	GLA	Pan-ethnic	≤1 in 500	99%	Reduced
Factor IX deficiency/hemophilia B (XL) NM_000133.3	F9	Pan-ethnic	≤1 in 500	99%	Reduced
Factor V Leiden (AD) NM_000130.4	F5*	Pan-ethnic	1 in 26	99%	1 in 2,500
Factor XI deficiency/hemophilia C (AR) NM_000128.3	F11	Ashkenazi Jewish	1 in 11	99%	1 in 1,000
		Pan-ethnic	≤1 in 500	99%	Reduced
Familial dysautonomia (AR) NM_003640.3	ELP1	Ashkenazi Jewish	1 in 36	99%	1 in 3,500
		Pan-ethnic	≤1 in 500	99%	Reduced

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Familial hypercholesterolemia (AD) NM_000527.4	LDLR	Afrikaner	1 in 72	99%	1 in 7,100
		Ashkenazi Jewish	1 in 69	99%	1 in 6,800
		French Canadian	1 in 270	99%	1 in 26,900
		Pan-ethnic	1 in 250	99%	1 in 24,900
Familial hypercholesterolemia (AR) NM_015627.2	LDLRAP1	Pan-ethnic	≤1 in 500	99%	Reduced
		Sardinian	1 in 143	99%	1 in 14,200
Familial hyperinsulinism (AR) NM_000352.4	ABCC8	Ashkenazi Jewish	1 in 52	99%	1 in 5,100
		Finnish	1 in 100	99%	1 in 9,900
		Pan-ethnic	1 in 177	99%	1 in 17,600
Familial hyperinsulinism (AR) NM_000525.3	KCNJ11	Pan-ethnic	≤1 in 500	99%	Reduced
Familial Mediterranean fever (AR) NM_000243.2	MEFV	Armenian	1 in 8	90%	1 in 71
		Ashkenazi Jewish	1 in 13	90%	1 in 121
		Pan-ethnic	1 in 64	90%	1 in 631
		Sephardic Jewish	1 in 14	90%	1 in 131
		Turkish	1 in 8	90%	1 in 71
Fanconi anemia type A (AR) NM_000135.2	FANCA	Afrikaner	1 in 83	99%	1 in 8,200
		Pan-ethnic	1 in 345	99%	1 in 34,400
		Sephardic Jewish	1 in 133	99%	1 in 13,200
		Spanish Roma	1 in 64	99%	1 in 6,300
Fanconi anemia type C (AR) NM_000136.2	FANCC	Ashkenazi Jewish	1 in 89	99%	1 in 8,800
		Pan-ethnic	1 in 417	99%	1 in 41,600
Fanconi anemia type G (AR) NM_004629.1	FANCG	African-American	1 in 100	99%	1 in 9,900
		Pan-ethnic	≤1 in 500	99%	Reduced
FKRP-related disorders (including Walker-Warburg syndrome) (AR) NM_024301.4	FKRP	Norwegian	1 in 116	99%	1 in 11,500
		Pan-ethnic	1 in 158	99%	1 in 15,700
FKTN-related disorders (including Walker-Warburg syndrome) (AR) NM_001079802.1	FKTN	Ashkenazi Jewish	1 in 80	99%	1 in 7,900
		Japanese	1 in 188	99%	1 in 18,700
		Pan-ethnic	≤1 in 500	99%	Reduced
Fragile X syndrome (XL) NM_002024.5	FMR1*	Ashkenazi Jewish	1 in 58	99%	1 in 5,700
		Asian	≤1 in 500	99%	Reduced
		Caucasian	1 in 187	99%	1 in 18,600
		Hispanic	≤1 in 500	99%	Reduced
		Pan-ethnic	1 in 259	99%	1 in 25,800
Fumarate hydratase deficiency (AR) NM_000143.3	FH	Pan-ethnic	≤1 in 500	99%	Reduced
Galactokinase deficiency galactosemia (AR) NM_000154.1	GALK1	Pan-ethnic	1 in 122	99%	1 in 12,100
		Roma	1 in 47	99%	1 in 4,600
Galactosemia (GALT-related) (AR) NM_000155.3	GALT	African-American	1 in 87	99%	1 in 8,600
		Ashkenazi Jewish	1 in 156	99%	1 in 15,500
		Irish Traveller	1 in 11	99%	1 in 1,000
		Pan-ethnic	1 in 100	99%	1 in 9,900
Gaucher disease (AR) NM_001005741.2	GBA*	Ashkenazi Jewish	1 in 15	94%	1 in 234
		Pan-ethnic	1 in 158	72%	1 in 561
Gitelman syndrome (SLC12A3-related) (AR) NM_000339.2	SLC12A3	Pan-ethnic	1 in 100	99%	1 in 9,900

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GJB2-related DFNB1 nonsyndromic hearing loss and deafness (AR) NM_004004.5	GJB2	Ashkenazi Jewish	1 in 13	99%	1 in 1,200
		Pan-ethnic	1 in 50	99%	1 in 4,900
		Thai	1 in 9	99%	1 in 800
Glucose-6-phosphate dehydrogenase deficiency (XL) NM_001042351.2	G6PD	Pan-ethnic	1 in 10	99%	1 in 900
Glutaric acidemia type I (AR) NM_000159.3	GCDH	Amish	1 in 9	99%	1 in 800
		Oji-Cree First Nations	1 in 9	99%	1 in 800
		Pan-ethnic	1 in 87	99%	1 in 8,600
Glutaric acidemia type II (AR) NM_000126.3	ETFA	Pan-ethnic	≤1 in 500	99%	Reduced
Glutaric acidemia type II (AR) NM_004453.3	ETFDH	Asian	1 in 87	99%	1 in 8,600
		Pan-ethnic	1 in 250	99%	1 in 24,900
Glycine encephalopathy (AR) NM_000481.3	AMT	Finnish	1 in 142	99%	1 in 14,100
		Pan-ethnic	1 in 325	99%	1 in 32,400
Glycine encephalopathy (AR) NM_000170.2	GLDC	Caucasian	1 in 141	99%	1 in 14,000
		Pan-ethnic	1 in 165	99%	1 in 16,400
Glycogen storage disease type Ia (AR) NM_000151.3	G6PC	Ashkenazi Jewish	1 in 71	95%	1 in 1,400
		Pan-ethnic	1 in 177	95%	1 in 3,520
Glycogen storage disease type Ib (AR) NM_001164277.1	SLC37A4	Pan-ethnic	1 in 354	95%	1 in 7,060
Glycogen storage disease type II (Pompe disease) (AR) NM_000152.3	GAA	African-American	1 in 60	99%	1 in 5,900
		Ashkenazi Jewish	1 in 58	99%	1 in 5,700
		Asian	1 in 112	99%	1 in 11,100
		Pan-ethnic	1 in 100	99%	1 in 9,900
Glycogen storage disease type III (AR) NM_000642.2	AGL	Faroese	1 in 28	95%	1 in 540
		Pan-ethnic	1 in 159	95%	1 in 3,160
		Sephardic Jewish (Moroccan)	1 in 34	95%	1 in 660
Glycogen storage disease type IV/adult polyglucosan body disease (AR) NM_000158.3	GBE1	Ashkenazi Jewish	1 in 68	99%	1 in 6,700
		Pan-ethnic	1 in 387	99%	1 in 38,600
Glycogen storage disease type V (AR) NM_005609.3	PYGM	Caucasian	1 in 158	99%	1 in 15,700
		Sephardic Jewish (Kurdish)	1 in 84	99%	1 in 8,300
Glycogen storage disease type VII (AR) NM_000289.5	PFKM	Ashkenazi Jewish	1 in 250	99%	1 in 24,900
		Pan-ethnic	≤1 in 500	99%	Reduced
GRACILE syndrome/BCS1L-related disorders (AR) NM_004328.4	BCS1L	Caucasian	1 in 407	99%	1 in 40,600
		Finnish	1 in 108	99%	1 in 10,700
		Pan-ethnic	≤1 in 500	99%	Reduced
Guanidinoacetate methyltransferase deficiency (AR) NM_000156.5	GAMT	Pan-ethnic	≤1 in 500	99%	Reduced
		Portuguese	1 in 125	99%	1 in 12,400
HBB-related hemoglobinopathies (AR) NM_000518.4	HBB	African-American	1 in 8	99%	1 in 700
		Asian	1 in 54	99%	1 in 5,300
		Caucasian	1 in 373	99%	1 in 37,200
		Hispanic	1 in 17	99%	1 in 1,600
		Mediterranean	1 in 28	99%	1 in 2,700
		Pan-ethnic	1 in 49	99%	1 in 4,800

Carrier screening gene list and carrier detection rates

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	DETECTION RATE	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Hereditary fructose intolerance (AR) NM_000035.3	ALDOB	African-American	1 in 226	99%	1 in 22,500
		Middle Eastern	1 in 97	99%	1 in 9,600
		Pan-ethnic	1 in 122	99%	1 in 12,100
Hereditary hemochromatosis (HFE-related) (AR) NM_000410.3	HFE	African-American	1 in 16	99%	1 in 1,500
		Asian	1 in 11	99%	1 in 1,000
		Hispanic	1 in 4	99%	1 in 300
		Northern European	1 in 3	99%	1 in 200
Hereditary hemochromatosis (HJV-related) (AR) NM_213653.3	HJV	Pan-ethnic	≤1 in 500	99%	Reduced
Hereditary hemochromatosis (TFR2-related) (AR) NM_003227.3	TFR2	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome (AR) NM_000195.4	HPS1	Pan-ethnic	≤1 in 500	99%	Reduced
		Puerto Rican (Northwestern)	1 in 21	99%	1 in 2,000
Hermansky-Pudlak syndrome (AR) NM_032383.4	HPS3	Ashkenazi Jewish	1 in 235	99%	1 in 23,400
		Pan-ethnic	≤1 in 500	99%	Reduced
		Puerto Rican (Central)	1 in 63	99%	1 in 6,200
Holocarboxylase synthetase deficiency (AR) NM_000411.6	HLCS	Faroese	1 in 20	99%	1 in 1,900
		Japanese	1 in 158	99%	1 in 15,700
		Pan-ethnic	1 in 224	99%	1 in 22,300
Homocystinuria (AR) NM_000071.2	CBS	Norwegian	1 in 40	99%	1 in 3,900
		Pan-ethnic	1 in 224	99%	1 in 22,300
		Qatari	1 in 21	99%	1 in 2,000
Homocystinuria due to MTHFR deficiency (AR) NM_005957.4	MTHFR	Sephardic Jewish (Bukharian)	1 in 39	99%	1 in 3,800
Homocystinuria, cobalamin E type (AR) NM_002454.2	MTRR	Pan-ethnic	≤1 in 500	99%	Reduced
Hydrolethalus syndrome type 1 (AR) NM_145014.2	HYLS1	Finnish	1 in 40	99%	1 in 3,900
		Pan-ethnic	≤1 in 500	99%	Reduced
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (AR) NM_014252.3	SLC25A15	Metis (Saskatchewan)	1 in 19	99%	1 in 1,800
		Pan-ethnic	≤1 in 500	99%	Reduced
Hypohidrotic ectodermal dysplasia (XL) NM_001399.4	EDA	Pan-ethnic	1 in 112	99%	1 in 11,100
Hypophosphatasia (AR) NM_000478.5	ALPL	Mennonite	1 in 25	95%	1 in 480
		Pan-ethnic	1 in 150	95%	1 in 2,980
Inclusion body myopathy 2 (AR) NM_001128227.2	GNE	Pan-ethnic	1 in 179	99%	1 in 17,800
		Sephardic Jewish (Iranian)	1 in 10	99%	1 in 900
Isovaleric acidemia (AR) NM_002225.3	IVD	Pan-ethnic	1 in 250	99%	1 in 24,900
Joubert syndrome 2/TMEM216-related disorders (AR) NM_001173990.2	TMEM216	Ashkenazi Jewish	1 in 92	99%	1 in 9,100
		Pan-ethnic	≤1 in 500	99%	Reduced
Junctional epidermolysis bullosa (AR) NM_000227.4	LAMA3	Pan-ethnic	≤1 in 500	99%	Reduced

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Junctional epidermolysis bullosa (AR) NM_000228.2	LAMB3	Pan-ethnic	1 in 317	99%	1 in 31,600
Junctional epidermolysis bullosa (AR) NM_005562.2	LAMC2	Pan-ethnic	≤1 in 500	99%	Reduced
Krabbe disease (AR) NM_000153.3	GALC	Druze	1 in 6	99%	≤1 in 500
		Pan-ethnic	1 in 158	99%	1 in 15,700
LAMA2-related muscular dystrophy (AR) NM_000426.3	LAMA2	Pan-ethnic	1 in 87	99%	1 in 8,600
Leber congenital amaurosis 2 (AR) NM_000329.2	RPE65	Pan-ethnic	1 in 228	99%	1 in 22,700
		Sephardic Jewish	1 in 90	99%	1 in 8,900
Leber congenital amaurosis 5 (AR) NM_181714.3	LCA5	Pan-ethnic	1 in 645	97%	Reduced
Leber congenital amaurosis 8/CRB1-related disorders (AR) NM_201253.2	CRB1	Pan-ethnic	1 in 112	99%	1 in 11,100
Leber congenital amaurosis 10/CEP290-related disorders (AR) NM_025114.3	CEP290	Pan-ethnic	1 in 185	99%	1 in 18,400
Leber congenital amaurosis 13 (AR) NM_152443.2	RDH12	Pan-ethnic	1 in 460	99%	1 in 45,900
Leigh syndrome, French Canadian type (AR) NM_133259.3	LRPPRC	French Canadian (Saguenay-Lac-St-Jean)	1 in 23	99%	1 in 2,200
		Pan-ethnic	≤1 in 500	99%	Reduced
Lethal congenital contracture syndrome 1/lethal arthrogryposis with anterior horn cell disease (AR) NM_001003722.1	GLE1	Finnish	1 in 100	99%	1 in 9,900
		Pan-ethnic	≤1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (AR) NM_003907.2	EIF2B5	Pan-ethnic	≤1 in 500	99%	Reduced
Limb-girdle muscular dystrophy type 2A (calpainopathy) (AR) NM_000070.2s	CAPN3	Pan-ethnic	1 in 134	99%	1 in 13,300
Limb-girdle muscular dystrophy type 2B (dysferlinopathy) (AR) NM_003494.3	DYSF	Pan-ethnic	1 in 311	99%	1 in 31,000
		Sephardic Jewish (Libyan)	1 in 10	99%	1 in 900
Limb-girdle muscular dystrophy type 2C (AR) NM_000231.2	SGCG	Caucasian	1 in 571	99%	Reduced
		Japanese	1 in 374	99%	1 in 37,300
		Moroccan	1 in 250	99%	1 in 24,900
		Pan-ethnic	≤1 in 500	99%	Reduced
		Roma	1 in 59	99%	1 in 5,800
Limb-girdle muscular dystrophy type 2D (AR) NM_000023.2	SGCA	Caucasian	1 in 286	99%	1 in 28,500
		Finnish	1 in 150	99%	1 in 14,900
		Pan-ethnic	≤1 in 500	99%	Reduced
Limb-girdle muscular dystrophy type 2E (AR) NM_000232.4	SGCB	Caucasian	1 in 404	92%	1 in 5,038
		Pan-ethnic	≤1 in 500	92%	Reduced
Lipoid congenital adrenal hyperplasia (STAR-related) (AR) NM_000349.2	STAR	Korean	1 in 170	99%	1 in 16,900
		Pan-ethnic	≤1 in 500	99%	Reduced
Lipoprotein lipase deficiency (AR) NM_000237.2	LPL	French Canadian (Saguenay-Lac-St-Jean)	1 in 46	99%	1 in 4,500
		Pan-ethnic	≤1 in 500	99%	Reduced
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (AR) NM_000182.4	HADHA	Caucasian	1 in 250	99%	1 in 24,900
		Finnish	1 in 125	99%	1 in 12,400
		Pan-ethnic	1 in 350	99%	1 in 34,900

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Lysinuric protein intolerance (AR) NM_001126106.2	SLC7A7	Finnish	1 in 120	95%	1 in 2,380
		Japanese	1 in 120	95%	1 in 2,380
		Pan-ethnic	≤1 in 500	99%	Reduced
Lysosomal acid lipase deficiency (AR) NM_000235.3	LIPA	Caucasian	1 in 112	94%	1 in 1,850
		Sephardic Jewish (Iranian)	1 in 33	94%	1 in 534
Major histocompatibility complex class II deficiency (CIITA-related) (AR) NM_000246.3	CIITA	Pan-ethnic	≤1 in 500	99%	Reduced
Maple syrup urine disease type 1a (AR) NM_000709.3	BCKDHA	Mennonite	1 in 10	99%	1 in 900
		Pan-ethnic	1 in 373	99%	1 in 37,200
Maple syrup urine disease type 1b (AR) NM_183050.2	BCKDHB	Ashkenazi Jewish	1 in 97	99%	1 in 9,600
		Pan-ethnic	1 in 346	99%	1 in 34,500
Maple syrup urine disease type 2 (AR) NM_001918.3	DBT	Pan-ethnic	≤1 in 500	99%	Reduced
Medium chain acyl-coa dehydrogenase deficiency (AR) NM_000016.5	ACADM	Northern European	1 in 40	99%	1 in 3,900
		Pan-ethnic	1 in 66	99%	1 in 6,500
Megalencephalic leukoencephalopathy with subcortical cysts type 1 (AR) NM_015166.3	MLC1	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish (Libyan)	1 in 40	99%	1 in 3,900
Menkes disease/ATP7A-related disorders (XL) NM_000052.6	ATP7A	Pan-ethnic	≤1 in 500	99%	Reduced
Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5	ARSA	Navajo	1 in 40	95%	1 in 780
		Pan-ethnic	1 in 100	95%	1 in 1,980
		Sephardic Jewish	1 in 46	95%	1 in 900
Methylmalonic acidemia (AR) NM_172250.2	MMAA	Pan-ethnic	1 in 316	97%	1 in 10,500
Methylmalonic acidemia (AR) NM_052845.3	MMAB	Pan-ethnic	1 in 456	98%	1 in 22,750
Methylmalonic acidemia (AR) NM_000255.3	MUT	Pan-ethnic	1 in 204	96%	1 in 5,075
Methylmalonic acidemia with homocystinuria, cobalamin C type (AR) NM_015506.2	MMACHC	Pan-ethnic	1 in 123	99%	1 in 12,200
Methylmalonic acidemia with homocystinuria, cobalamin D type (AR) NM_015702.2	MMADHC*	Pan-ethnic	≤1 in 500	99%	Reduced
Microphthalmia/clinical anophthalmia (VSX2-related) (AR) NM_182894.2	VSX2	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish	1 in 145	99%	1 in 14,400
Mitochondrial complex I deficiency/Leigh syndrome (AR) NM_024120.4	NDUFAF5	Ashkenazi Jewish	1 in 290	99%	1 in 28,900
		Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex I deficiency/Leigh syndrome (AR) NM_004553.4	NDUFS6	Ashkenazi Jewish	1 in 290	99%	1 in 28,900
		Caucasus Jewish	1 in 24	99%	1 in 2,300
		Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial DNA depletion syndrome (MPV17-related) (AR) NM_002437.4	MPV17	Navajo	1 in 20	96%	1 in 475
		Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial myopathy and sideroblastic anemia 1 (AR) NM_025215.5	PUS1	Pan-ethnic	≤1 in 500	99%	Reduced

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Mitochondrial neurogastrointestinal encephalopathy disease (AR) NM_001953.4	TYMP	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish	1 in 158	99%	1 in 15,700
MKS1-related disorders (AR) NM_017777.3	MKS1	Finnish	1 in 47	95%	1 in 920
		Pan-ethnic	1 in 260	95%	1 in 5,180
Mucolipidosis type II/III (AR) NM_024312.4	GNPTAB	Irish Traveller	1 in 15	99%	1 in 1,400
		Pan-ethnic	1 in 200	99%	1 in 19,900
Mucolipidosis type III (AR) NM_032520.4	GNPTG	Pan-ethnic	≤1 in 500	99%	Reduced
Mucolipidosis type IV (AR) NM_020533.2	MCOLN1	Ashkenazi Jewish	1 in 100	99%	1 in 9,900
		Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolysaccharidosis type I (AR) NM_000203.4	IDUA	Pan-ethnic	1 in 148	97%	1 in 4,900
Mucopolysaccharidosis type II (Hunter syndrome) (XL) NM_000202.6	IDS	Pan-ethnic	≤1 in 500	90%	Reduced
Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome) (AR) NM_000199.3	SGSH	Northern European	1 in 173	99%	1 in 17,200
		Pan-ethnic	1 in 215	99%	1 in 21,400
		Taiwanese	≤1 in 500	99%	Reduced
Mucopolysaccharidosis type IIIB (AR) NM_000263.3	NAGLU	Pan-ethnic	1 in 224	99%	1 in 22,300
Mucopolysaccharidosis type IIIC (Sanfilippo syndrome)/ Retinitis pigmentosa 73 (AR) NM_152419.2	HGSNAT	Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolysaccharidosis type IIID (Sanfilippo syndrome) (AR) NM_002076.3	GNS	Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolysaccharidosis type IVB (Morquio B syndrome)/ GM1 gangliosidosis (AR) NM_000404.2	GLB1	Pan-ethnic	1 in 158	99%	1 in 15,700
		Roma	1 in 50	99%	1 in 4,900
		South Brazilian	1 in 58	99%	1 in 5,700
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome) (AR) NM_000046.3	ARSB	Pan-ethnic	1 in 250	99%	1 in 24,900
Mucopolysaccharidosis type IX (AR) NM_153281.1	HYAL1	Pan-ethnic	≤1 in 500	99%	Reduced
Multiple sulfatase deficiency (AR) NM_182760.3	SUMF1	Pan-ethnic	≤1 in 500	99%	Reduced
N-acetylglutamate synthase deficiency (AR) NM_153006.2	NAGS	Pan-ethnic	≤1 in 500	99%	Reduced
Nemaline myopathy 2 (AR) NM_001271208.1	NEB*	Ashkenazi Jewish	1 in 108	99%	1 in 10,700
		Pan-ethnic	1 in 158	95%	1 in 3,140
Nephrogenic diabetes insipidus (AR) NM_000486.5	AQP2	Pan-ethnic	1 in 1118	99%	Reduced
Nephrotic syndrome/congenital Finnish nephrosis (AR) NM_004646.3	NPHS1	Finnish	1 in 46	99%	1 in 4,500
		Old Order Mennonite	1 in 12	99%	1 in 1,100
		Pan-ethnic	≤1 in 500	99%	Reduced
Nephrotic syndrome/steroid-resistant nephrotic syndrome (AR) NM_014625.3	NPHS2	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid-lipofuscinosis (AR) NM_001042432.1	CLN3	Pan-ethnic	1 in 230	99%	1 in 22,900

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Neuronal ceroid-lipofuscinosis (AR) NM_006493.2	CLN5	Finnish	1 in 115	99%	1 in 11,400
		Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid-lipofuscinosis (AR) NM_017882.2	CLN6	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid-lipofuscinosis (AR) NM_152778.2	MFSD8	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid-lipofuscinosis (AR) NM_000310.3	PPT1	Finnish	1 in 70	98%	1 in 3,450
		Pan-ethnic	1 in 199	98%	1 in 9,900
Neuronal ceroid-lipofuscinosis (AR) NM_000391.3	TPP1	Newfoundland	1 in 53	97%	1 in 1,734
		Pan-ethnic	1 in 250	97%	1 in 8,300
Neuronal ceroid-lipofuscinosis/Northern epilepsy (AR) NM_018941.3	CLN8	Finnish	1 in 135	99%	1 in 13,400
		Pan-ethnic	≤1 in 500	99%	Reduced
Niemann-pick disease type A/B (AR) NM_000543.4	SMPD1	Ashkenazi Jewish	1 in 90	95%	1 in 1,780
		Pan-ethnic	1 in 250	95%	1 in 4,980
Niemann-pick disease type C (AR) NM_000271.4	NPC1	Pan-ethnic	1 in 183	99%	1 in 18,200
Niemann-pick disease type C (AR) NM_006432.3	NPC2	Pan-ethnic	1 in 871	99%	Reduced
Nijmegen breakage syndrome (AR) NM_002485.4	NBN*	Eastern European	1 in 155	99%	1 in 15,400
		Pan-ethnic	≤1 in 500	99%	Reduced
Ornithine aminotransferase deficiency (AR) NM_000274.3	OAT	Finnish	1 in 126	99%	1 in 12,500
		Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish	1 in 177	99%	1 in 17,600
Ornithine transcarbamylase deficiency (XL) NM_000531.5	OTC	Pan-ethnic	≤1 in 500	85%	Reduced
Osteopetrosis (AR) NM_006019.3	TCIRG1	Ashkenazi Jewish	1 in 350	99%	1 in 34,900
		Chuvash	1 in 30	99%	1 in 2,900
		Pan-ethnic	1 in 317	99%	1 in 31,600
Pendred syndrome (AR) NM_000441.1	SLC26A4	Asian	1 in 74	99%	1 in 7,300
		Pan-ethnic	1 in 80	99%	1 in 7,900
Peroxisomal acyl-coa oxidase deficiency (AR) NM_004035.6	ACOX1	Pan-ethnic	≤1 in 500	99%	Reduced
Phenylalanine hydroxylase deficiency (AR) NM_000277.1	PAH	African-American	1 in 111	99%	1 in 11,000
		Ashkenazi Jewish East	1 in 225	99%	1 in 22,400
		East Asian	1 in 50	96%	1 in 1,225
		Finnish	1 in 225	99%	1 in 22,400
		Irish	1 in 33	99%	1 in 3,200
		Japanese	1 in 200	99%	1 in 19,900
		Pan-ethnic	1 in 58	99%	1 in 5,700
		Turkish	1 in 26	99%	1 in 2,500
Phosphoglycerate dehydrogenase deficiency/Neu-Laxova syndrome (AR) NM_006623.3	PHGDH	Ashkenazi Jewish	1 in 400	99%	1 in 39,900
		Pan-ethnic	≤1 in 500	99%	Reduced
Polycystic kidney disease (AR) NM_138694.3	PKHD1	Pan-ethnic	1 in 70	99%	1 in 6,900
Polymicrogyria (AR) NM_005682.6	ADGRG1	Pan-ethnic	≤1 in 500	99%	Reduced

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POMGNT1-related disorders (AR) NM_017739.3	POMGNT1	Finnish	1 in 111	99%	1 in 11,000
		Pan-ethnic	≤1 in 500	99%	Reduced
Pontocerebellar hypoplasia (AR) NM_020320.3	RARS2	Pan-ethnic	≤1 in 500	99%	Reduced
Pontocerebellar hypoplasia (AR) NM_016955.3	SEPSECS	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish (Moroccan and Iraqi)	1 in 43	99%	1 in 4,200
Pontocerebellar hypoplasia (AR) NM_003384.2	VRK1	Ashkenazi Jewish	1 in 225	99%	1 in 22,400
		Pan-ethnic	≤1 in 500	99%	Reduced
Postnatal progressive microcephaly with seizures and brain atrophy/infantile cerebral and cerebellar atrophy (AR) NM_004268.4	MED17	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish	1 in 20	99%	1 in 1,900
Primary carnitine deficiency (AR) NM_003060.3	SLC22A5	Faroese	1 in 9	99%	1 in 800
		Japanese	1 in 100	99%	1 in 9,900
		Pan-ethnic	1 in 71	99%	1 in 7,000
Primary ciliary dyskinesia (AR) NM_001369.2	DNAH5	Pan-ethnic	1 in 109	99%	1 in 10,800
Primary ciliary dyskinesia (AR) NM_012144.3	DNAI1	Pan-ethnic	1 in 250	99%	1 in 24,900
Primary ciliary dyskinesia (AR) NM_023036.4	DNAI2	Ashkenazi Jewish	1 in 200	99%	1 in 19,900
		Pan-ethnic	1 in 354	99%	1 in 35,300
Primary hyperoxaluria type 1 (AR) NM_000030.2	AGXT	Pan-ethnic	1 in 135	99%	1 in 13,400
Primary hyperoxaluria type 2 (AR) NM_012203.1	GRHPR	Pan-ethnic	≤1 in 500	99%	Reduced
Primary hyperoxaluria type 3 (AR) NM_138413.3	HOGA1	Pan-ethnic	1 in 354	99%	1 in 35,300
Progressive familial intrahepatic cholestasis type 2 (AR) NM_003742.2	ABCB11	Pan-ethnic	1 in 100	99%	1 in 9,900
Propionic acidemia (AR) NM_000282.3	PCCA	Arab	1 in 100	96%	1 in 2,475
		Pan-ethnic	1 in 224	96%	1 in 5,575
Propionic acidemia (AR) NM_000532.4	PCCB	Arab	1 in 100	99%	1 in 9,900
		Greenlandic Inuit	1 in 20	99%	1 in 1,900
		Pan-ethnic	1 in 224	99%	1 in 22,300
Prothrombin-related thrombophilia (AD) NM_000506.3	F2*	Pan-ethnic	1 in 62	99%	1 in 6,100
PRPS1-related disorders (XL) NM_002764.3	PRPS1	Pan-ethnic	≤1 in 500	99%	Reduced
PSAP-related disorders (AR) NM_002778.3	PSAP	Pan-ethnic	≤1 in 500	99%	Reduced
Pycnodysostosis (AR) NM_000396.3	CTSK	Pan-ethnic	1 in 438	99%	1 in 43,700
Pyruvate carboxylase deficiency (AR) NM_000920.3	PC	Algonquian Indian	1 in 10	95%	1 in 180
		Pan-ethnic	1 in 250	95%	1 in 4,980
Pyruvate dehydrogenase deficiency (AR) NM_000925.3	PDHB	Pan-ethnic	≤1 in 500	99%	Reduced
Pyruvate dehydrogenase deficiency (XL) NM_000284.3	PDHA1	Pan-ethnic	≤1 in 500	99%	Reduced

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Renal tubular acidosis with deafness (AR) NM_001692.3	ATP6V1B1	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish	1 in 140	99%	1 in 13,900
Retinitis pigmentosa 25 (AR) NM_001142800.1	EYS	Pan-ethnic	1 in 129	99%	1 in 12,800
		Sephardic Jewish	1 in 42	99%	1 in 4,100
Retinitis pigmentosa 26 (AR) NM_001030311.2	CERKL	Pan-ethnic	1 in 137	99%	1 in 13,600
		Sephardic Jewish	1 in 24	99%	1 in 2,300
Retinitis pigmentosa 28 (AR) NM_001201543.1	FAM161A	Ashkenazi Jewish	1 in 214	99%	1 in 21,300
		Pan-ethnic	1 in 289	99%	1 in 28,800
		Sephardic Jewish	1 in 41	99%	1 in 4,000
Rhizomelic chondrodysplasia punctata type 1/Refsum disease (AR) NM_000288.3	PEX7	Pan-ethnic	1 in 157	99%	1 in 15,600
Rhizomelic chondrodysplasia punctata type 3 (AR) NM_003659.3	AGPS	Pan-ethnic	≤1 in 500	99%	Reduced
Roberts syndrome (AR) NM_001017420.2	ESCO2	Pan-ethnic	≤1 in 500	99%	Reduced
RPGRIP1L-related disorders (AR) NM_015272.2	RPGRIP1L*	Pan-ethnic	1 in 259	95%	1 in 5,160
RTEL1-related disorders (AR) NM_032957.4	RTEL1	Ashkenazi Jewish	1 in 222	99%	1 in 22,100
		Pan-ethnic	≤1 in 500	99%	Reduced
Sandhoff disease (AR) NM_000521.3	HEXB	Metis (Saskatchewan)	1 in 15	99%	1 in 1,400
		Pan-ethnic	1 in 180	99%	1 in 17,900
Schimke immuno-osseous dysplasia (AR) NM_014140.3	SMARCAL1	Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency (AR) NM_001033855.2	DCLRE1C	Navajo and Apache	1 in 10	99%	1 in 900
		Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency/Omenn syndrome (AR) NM_000536.3	RAG2	Pan-ethnic	≤1 in 500	99%	Reduced
Severe congenital neutropenia (AR) NM_007259.4	VPS45	Pan-ethnic	≤1 in 500	99%	Reduced
Sialic acid storage disorders (AR) NM_012434.4	SLC17A5	Finnish	1 in 100	99%	1 in 9,900
		Pan-ethnic	≤1 in 500	99%	Reduced
Sjögren-Larsson syndrome (AR) NM_000382.2	ALDH3A2	Pan-ethnic	≤1 in 500	99%	Reduced
		Swedish	1 in 250	99%	1 in 24,900
SLC26A2-related disorders (AR) NM_000112.3	SLC26A2	Finnish	1 in 75	95%	1 in 1,480
		Pan-ethnic	1 in 158	95%	1 in 3,140
SLC35A3-related disorder (AR) NM_012243.2	SLC35A3	Ashkenazi Jewish	1 in 469	99%	1 in 46,800
		Pan-ethnic	≤1 in 500	99%	Reduced
Smith-Lemli-Opitz syndrome (AR) NM_001360.2	DHCR7	African-American	1 in 339	96%	1 in 8,450
		Ashkenazi Jewish	1 in 41	96%	1 in 1,000
		Hispanic	1 in 135	96%	1 in 3,350
		Northern European	1 in 50	96%	1 in 1,225
		Pan-ethnic	1 in 71	96%	1 in 1,750
		Sephardic Jewish	1 in 68	96%	1 in 1,675
		Southern European	1 in 83	96%	1 in 2,050

Carrier screening gene list and carrier detection rates

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	DETECTION RATE	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Spastic paraplegia type 15 (AR) NM_015346.3	ZFYVE26	Pan-ethnic	≤1 in 500	99%	Reduced
Spastic paraplegia type 49 (AR) NM_014844.3	TECPR2	Sephardic Jewish (Bukharian)	1 in 38	99%	1 in 3,700
Spinal muscular atrophy (AR) NM_000344.3	SMN1*	African-American	1 in 66	72%	1 in 233
		Ashkenazi Jewish	1 in 41	94%	1 in 667
		Asian	1 in 53	93%	1 in 743
		Caucasian	1 in 35	94%	1 in 567
		Hispanic	1 in 117	90%	1 in 1,161
Spondylothoracic dysostosis (AR) NM_001039958.1	MESP2	Pan-ethnic	1 in 224	99%	1 in 22,300
		Puerto Rican	1 in 55	99%	1 in 5,400
Steel syndrome (AR) NM_032888.3	COL27A1*	Pan-ethnic	≤1 in 500	99%	Reduced
		Puerto Rican	1 in 51	99%	1 in 5,000
Stüve-Wiedemann syndrome (AR) NM_002310.5	LIFR	Pan-ethnic	≤1 in 500	99%	Reduced
Tay-Sachs disease/hexosaminidase A deficiency (AR) NM_000520.4	HEXA	Ashkenazi Jewish	1 in 27	99%	1 in 2,600
		Asian	1 in 126	99%	1 in 12,500
		Caucasian	1 in 182	99%	1 in 18,100
		French Canadian	1 in 27	99%	1 in 2,600
		Irish	1 in 41	99%	1 in 4,000
		Pan-ethnic	1 in 250	99%	1 in 24,900
		Sephardic Jewish	1 in 125	99%	1 in 12,400
Tetrahydrobiopterin deficiency (AR) NM_000317.2	PTS	Chinese	1 in 122	99%	1 in 12,100
		Pan-ethnic	1 in 433	99%	1 in 43,200
Transient infantile liver failure (AR) NM_018006.4	TRMU	Pan-ethnic	≤1 in 500	99%	Reduced
		Sephardic Jewish (Yemenite)	1 in 34	99%	1 in 3,300
Tyrosine hydroxylase deficiency (AR) NM_199292.2	TH	Caucasian	1 in 224	99%	1 in 22,300
		Pan-ethnic	≤1 in 500	99%	Reduced
Tyrosinemia type I (AR) NM_000137.2	FAH	Ashkenazi Jewish	1 in 143	95%	1 in 2,840
		French Canadian	1 in 66	95%	1 in 1,300
		French Canadian (Saguenay–Lac-St-Jean)	1 in 16	95%	1 in 300
		Pan-ethnic	1 in 125	95%	1 in 2,480
Tyrosinemia type II (AR) NM_000353.2	TAT	Pan-ethnic	1 in 250	99%	1 in 24,900
Usher syndrome type IB/MYO7A-related disorders (AR) NM_000260.3	MYO7A	Pan-ethnic	1 in 200	95%	1 in 3,980
Usher syndrome type IC/USH1C-related disorders (AR) NM_005709.3	USH1C*	French Canadian/ Acadian	1 in 227	99%	1 in 22,600
		Pan-ethnic	1 in 353	90%	1 in 3,521
		Sephardic Jewish	1 in 125	90%	1 in 1,241
Usher syndrome type ID (AR) NM_022124.5	CDH23	Pan-ethnic	1 in 202	95%	1 in 4,020
Usher syndrome type IF/PCDH15-related disorders (AR) NM_033056.3	PCDH15	Ashkenazi Jewish	1 in 78	99%	1 in 7,700
		Pan-ethnic	1 in 400	99%	1 in 39,900

Carrier screening gene list and carrier detection rates

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY BEFORE SCREENING	DETECTION RATE	CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
Usher syndrome type IIA/USH2A-related disorders (AR) NM_206933.2	USH2A	Caucasian	1 in 70	99%	1 in 6,900
		Pan-ethnic	1 in 112	99%	1 in 11,100
		Sephardic Jewish	1 in 36	99%	1 in 3,500
Usher syndrome type IIIA (AR) NM_174878.2	CLRN1	Ashkenazi Jewish	1 in 120	99%	1 in 11,900
		Pan-ethnic	1 in 533	99%	Reduced
Very long-chain acyl-coa dehydrogenase deficiency (AR) NM_000018.3	ACADVL	Pan-ethnic	1 in 100	99%	1 in 9,900
Wilson disease (AR) NM_000053.3	ATP7B	Ashkenazi Jewish	1 in 67	98%	1 in 3,300
		Canary Islander	1 in 25	98%	1 in 1,200
		Pan-ethnic	1 in 90	98%	1 in 4,450
		Sardinian	1 in 50	98%	1 in 2,450
		Sephardic Jewish	1 in 65	98%	1 in 3,200
WNT10A-related disorders (AR) NM_025216.2	WNT10A	Pan-ethnic	1 in 305	99%	1 in 30,400
X-linked adrenoleukodystrophy (XL) NM_000033.3	ABCD1	Pan-ethnic	1 in 16,800	99%	Reduced
		Sephardic Jewish	≤1 in 500	99%	Reduced
X-linked creatine transporter deficiency (XL) NM_005629.3	SLC6A8	Pan-ethnic	≤1 in 500	99%	Reduced
X-linked juvenile retinoschisis (XL) NM_000330.3	RS1	Pan-ethnic	≤1 in 500	99%	Reduced
X-linked myotubular myopathy (XL) NM_000252.2	MTM1	Pan-ethnic	≤1 in 500	96%	Reduced
X-linked severe combined immunodeficiency (XL) NM_000206.2	IL2RG	Pan-ethnic	≤1 in 500	99%	Reduced
Xeroderma pigmentosum complementation group A (AR) NM_000380.3	XPA	Japanese	1 in 100	99%	1 in 9,900
		Pan-ethnic	1 in 1,667	99%	Reduced
Xeroderma pigmentosum complementation group C (AR) NM_004628.4	XPC	Pan-ethnic	1 in 763	99%	Reduced
		Tunisian	1 in 50	99%	1 in 4,900
Zellweger spectrum disorder (AR) NM_000466.2	PEX1	Pan-ethnic	1 in 144	99%	1 in 14,300
Zellweger spectrum disorder (AR) NM_000318.2	PEX2	Ashkenazi Jewish	1 in 227	99%	1 in 22,600
		Pan-ethnic	≤1 in 500	99%	Reduced
Zellweger spectrum disorder (AR) NM_000287.3	PEX6	French Canadian	1 in 55	99%	1 in 5,400
		Pan-ethnic	1 in 294	99%	1 in 29,300
		Sephardic Jewish	1 in 18	99%	1 in 1,700
Zellweger spectrum disorder (AR) NM_153818.1	PEX10	Pan-ethnic	1 in 606	94%	Reduced
Zellweger spectrum disorder (AR) NM_000286.2	PEX12	Pan-ethnic	1 in 409	99%	1 in 40,800

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