

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. For genes associated with both dominant and recessive inheritance, the numbers in this table apply to the recessive condition(s) associated with the gene, unless otherwise noted. 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 may vary based on the ethnic background(s) of an individual. For any genes marked with an asterisk\*, refer to the Limitations section of the patient report for detailed coverage information. In the case of a sample-specific limitation, "N/A" indicates that a residual risk value could not be calculated. AR = autosomal recessive, XL = X-linked, AD = autosomal dominant.

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
3-hydroxy-3-methylglutaryl-CoA lyase deficiency (AR) NM_000191.2	HMGCL	Pan-ethnic	≤1 in 500	99%	Reduced
17-beta hydroxysteroid dehydrogenase 3 deficiency (AR) NM_000197.1	HSD17B3	Pan-ethnic	≤1 in 500	99%	Reduced
ABCA3-related conditions (AR) NM_001089.2	ABCA3	Pan-ethnic	1 in 277	99%	1 in 27600
ABCA4-related conditions (AR) NM_000350.2	ABCA4	Pan-ethnic	1 in 45	90%	1 in 441
ABCB4-related conditions (AR) NM_000443.3	ABCB4	Pan-ethnic	1 in 204	99%	1 in 20300
ABCB11-related conditions (AR) NM_003742.2	ABCB11	Pan-ethnic	1 in 100	99%	1 in 9900
ABCC8-related conditions (AR) NM_000352.4 When the mother is a noncarrier, but the father is a carrier, there is a residual risk for focal disease (1 in 540 for the Ashkenazi Jewish population; undetermined in other ethnic groups)	ABCC8	Pan-ethnic	1 in 177	99%	1 in 17600
Abetalipoproteinemia (AR) NM_000253.3	MTTP	Pan-ethnic	≤1 in 500	99%	Reduced
Achromatopsia (CNGB3-related) (AR) NM_019098.4	CNGB3	Pan-ethnic	1 in 93	99%	1 in 9200
ACOX1-related conditions (AR) NM_004035.6	ACOX1	Pan-ethnic	≤1 in 500	99%	Reduced
Acrodermatitis enteropathica (AR) NM_130849.3	SLC39A4	Pan-ethnic	1 in 354	99%	1 in 35300
Adenosine deaminase deficiency (AR) NM_000022.2	ADA	Pan-ethnic	1 in 224	92%	1 in 2788
ADGRV1-related conditions (AR) NM_032119.3	ADGRV1	Pan-ethnic	1 in 223	99%	1 in 22200
AHI1-related conditions (AR) NM_017651.4	AHI1	Pan-ethnic	1 in 447	99%	1 in 44600
Aicardi-Goutieres syndrome 2 (AR) NM_024570.3	RNASEH2B	Pan-ethnic	≤1 in 500	99%	Reduced
Aicardi-Goutieres syndrome 3 (AR) NM_032193.3	RNASEH2C	Pan-ethnic	≤1 in 500	99%	Reduced
Aicardi-Goutieres syndrome 4 (AR) NM_006397.2	RNASEH2A	Pan-ethnic	≤1 in 500	99%	Reduced
Aicardi-Goutieres syndrome 5 (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
Alpha-mannosidosis (AR) NM_000528.3	MAN2B1	Pan-ethnic	1 in 354	99%	1 in 35300
Alpha-N-acetylgalactosaminidase deficiency (AR) NM_000262.2	NAGA	Pan-ethnic	≤1 in 500	99%	Reduced
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

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
Alport syndrome (COL4A3-related) (AR) NM_000091.4	COL4A3	Pan-ethnic	1 in 354	99%	1 in 35300
Alport syndrome (COL4A4-related) (AR) NM_000092.4	COL4A4	Pan-ethnic	1 in 353	99%	1 in 35200
Alström syndrome (AR) NM_015120.4	ALMS1	Pan-ethnic	≤1 in 500	99%	Reduced
Arginase deficiency (AR) NM_000045.3	ARG1	Pan-ethnic	1 in 274	99%	1 in 27300
Argininosuccinate lyase deficiency (AR) NM_000048.3	ASL	Pan-ethnic	1 in 133	90%	1 in 1321
ARL6-related conditions (AR) NM_177976.2	ARL6	Pan-ethnic	≤1 in 500	99%	Reduced
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
Aspartylglucosaminuria (AR) NM_000027.3	AGA	Pan-ethnic	≤1 in 500	99%	Reduced
Ataxia with vitamin E deficiency (AR) NM_000370.3	TTPA	Pan-ethnic	≤1 in 500	90%	Reduced
Ataxia-telangiectasia-like disorder (AR) NM_005591.3	MRE11	Pan-ethnic	≤1 in 500	99%	Reduced
ATM-related conditions (AR) NM_000051.3	ATM *	Pan-ethnic	1 in 100	99%	1 in 9900
ATP8B1-related conditions (AR) NM_005603.4	ATP8B1 *	Pan-ethnic	1 in 112	99%	1 in 11100
Atransferrinemia (AR) NM_001063.3	TF	Pan-ethnic	≤1 in 500	99%	Reduced
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia (AR) NM_000383.3	AIRE	Pan-ethnic	1 in 150	99%	1 in 14900
Autosomal recessive congenital ichthyosis (ABCA12-related) (AR) NM_173076.2	ABCA12	Pan-ethnic	≤1 in 500	99%	Reduced
Autosomal recessive congenital ichthyosis (TGM1-related) (AR) NM_000359.2	TGM1	Pan-ethnic	1 in 224	95%	1 in 4460
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (AR) NM_014363.5	SACS	Pan-ethnic	≤1 in 500	99%	Reduced
Bardet-Biedl syndrome (BBS7-related) (AR) NM_176824.2	BBS7	Pan-ethnic	≤1 in 500	99%	Reduced
Bardet-Biedl syndrome (BBS9-related) (AR) NM_198428.2	BBS9 *	Pan-ethnic	≤1 in 500	99%	Reduced
Bardet-Biedl syndrome (BBS10-related) (AR) NM_024685.3	BBS10	Pan-ethnic	1 in 354	99%	1 in 35300
Bardet-Biedl syndrome (BBS12-related) (AR) NM_152618.2	BBS12	Pan-ethnic	≤1 in 500	99%	Reduced
Barter syndrome type 1 (AR) NM_000338.2	SLC12A1	Pan-ethnic	1 in 224	99%	1 in 22300
Barter syndrome type 2 (AR) NM_000220.4	KCNJ1	Pan-ethnic	≤1 in 500	99%	Reduced
BBS1-related conditions (AR) NM_024649.4	BBS1	Pan-ethnic	1 in 330	99%	1 in 32900
BBS2-related conditions (AR) NM_031885.3	BBS2	Pan-ethnic	≤1 in 500	99%	Reduced
BBS4-related conditions (AR) NM_033028.4	BBS4	Pan-ethnic	≤1 in 500	99%	Reduced
BBS5-related conditions (AR) NM_152384.2	BBS5	Pan-ethnic	≤1 in 500	99%	Reduced
BCS1L-related conditions (AR) NM_004328.4	BCS1L	Pan-ethnic	≤1 in 500	99%	Reduced

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
Beta-ketothiolase deficiency (AR) NM_000019.3	ACAT1	Pan-ethnic	≤1 in 500	99%	Reduced
Beta-mannosidosis (AR) NM_005908.3	MANBA	Pan-ethnic	≤1 in 500	99%	Reduced
Biopterin-deficient hyperphenylalaninemia (PCBD1-related) (AR) NM_000281.3	PCBD1	Pan-ethnic	≤1 in 500	99%	Reduced
Biopterin-deficient hyperphenylalaninemia (PTS-related) (AR) NM_000317.2	PTS	Pan-ethnic	1 in 433	99%	1 in 43200
Biopterin-deficient hyperphenylalaninemia (QDPR-related) (AR) NM_000320.2	QDPR	Pan-ethnic	≤1 in 500	99%	Reduced
Biotin-responsive basal ganglia disease (AR) NM_025243.3	SLC19A3	Pan-ethnic	≤1 in 500	99%	Reduced
Biotinidase deficiency (AR) NM_000060.3	BTD	Pan-ethnic	1 in 125	99%	1 in 12400
Bloom syndrome (AR) NM_000057.3	BLM	Pan-ethnic	≤1 in 500	99%	Reduced
BRIP1-related conditions (AR) NM_032043.2	BRIP1	Pan-ethnic	≤1 in 500	99%	Reduced
Brittle cornea syndrome (PRDM5-related) (AR) NM_018699.3	PRDM5	Pan-ethnic	≤1 in 500	99%	Reduced
Brittle cornea syndrome (ZNF469-related) (AR) NM_001127464.2	ZNF469	Pan-ethnic	≤1 in 500	99%	Reduced
BSND-related conditions (AR) NM_057176.2	BSND	Pan-ethnic	≤1 in 500	99%	Reduced
Canavan disease (AR) NM_000049.2	ASPA	Pan-ethnic	1 in 159	99%	1 in 15800
Carbamoyl phosphate synthetase I deficiency (AR) NM_001875.4	CPS1	Pan-ethnic	≤1 in 500	99%	Reduced
Cardioencephalomyopathy (AR) NM_005138.2	SCO2	Pan-ethnic	1 in 387	99%	1 in 38600
Carnitine palmitoyltransferase I deficiency (AR) NM_001876.3	CPT1A	Pan-ethnic	≤1 in 500	99%	Reduced
Carnitine palmitoyltransferase II deficiency (AR) NM_000098.2	CPT2	Pan-ethnic	1 in 182	99%	1 in 18100
Carnitine-acylcarnitine translocase deficiency (AR) NM_000387.5	SLC25A20	Pan-ethnic	≤1 in 500	99%	Reduced
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	Pan-ethnic	≤1 in 500	99%	Reduced
Catecholaminergic polymorphic ventricular tachycardia (CASQ2-related) (AR) NM_001232.3	CASQ2	Pan-ethnic	1 in 224	99%	1 in 22300
CC2D2A-related conditions (AR) NM_001080522.2	CC2D2A	Pan-ethnic	1 in 426	99%	1 in 42500
CDH23-related conditions (AR) NM_022124.5	CDH23	Pan-ethnic	1 in 202	95%	1 in 4020
CEP290-related conditions (AR) NM_025114.3	CEP290	Pan-ethnic	1 in 185	99%	1 in 18400
Cerebellar ataxia, intellectual disability, and dysequilibrium syndrome 1 (AR) NM_003383.4	VLDLR	Pan-ethnic	≤1 in 500	99%	Reduced
Cerebral dysgenesis, neuropathy, ichthyosis, and keratoderma (AR) NM_004782.3	SNAP29	Pan-ethnic	≤1 in 500	99%	Reduced
Cerebrotendinous xanthomatosis (AR) NM_000784.3	CYP27A1	Pan-ethnic	1 in 112	98%	1 in 5550
CERKL-related conditions (AR) NM_001030311.2	CERKL	Pan-ethnic	1 in 137	99%	1 in 13600



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CFTR-related conditions (AR) NM_000492.3	CFTR *	Pan-ethnic - classic CF	1 in 45	99%	1 in 4400
		Pan-ethnic - classic CF and CFTR-related disorders	1 in 9	99%	1 in 800
Charcot-Marie-Tooth disease type 4D (AR) NM_006096.3	NDRG1	Pan-ethnic	≤1 in 500	99%	Reduced
Chediak-Higashi syndrome (AR) NM_000081.3	LYST	Pan-ethnic	≤1 in 500	99%	Reduced
Childhood-onset dystonia with optic atrophy and basal ganglia abnormalities (AR) NM_016011.3	MECR	Pan-ethnic	≤1 in 500	99%	Reduced
Chorea-acanthocytosis (AR) NM_033305.2	VPS13A *	Pan-ethnic	≤1 in 500	97%	Reduced
Chronic granulomatous disease (CYBA-related) (AR) NM_000101.3	CYBA	Pan-ethnic	≤1 in 500	99%	Reduced
Chronic granulomatous disease (NCF2-related) (AR) NM_000433.3	NCF2	Pan-ethnic	≤1 in 500	99%	Reduced
Citrin deficiency (AR) NM_014251.2	SLC25A13	Pan-ethnic	1 in 313	99%	1 in 31200
Citrullinemia type 1 (AR) NM_000050.4	ASS1	Pan-ethnic	1 in 120	96%	1 in 2975
CLN3-related conditions (AR) NM_001042432.1	CLN3	Pan-ethnic	1 in 230	99%	1 in 22900
CLRN1-related conditions (AR) NM_174878.2	CLRN1	Pan-ethnic	≤1 in 500	99%	Reduced
Cobalamin C deficiency (AR) NM_015506.2	MMACHC	Pan-ethnic	1 in 123	99%	1 in 12200
Cobalamin D deficiency (AR) NM_015702.2	MMADHC	Pan-ethnic	≤1 in 500	99%	Reduced
Cobalamin F deficiency (AR) NM_018368.3	LMBRD1	Pan-ethnic	≤1 in 500	99%	Reduced
Cockayne syndrome A (AR) NM_000082.3	ERCC8	Pan-ethnic	≤1 in 500	99%	Reduced
Cockayne syndrome B (AR) NM_000124.3	ERCC6	Pan-ethnic	1 in 377	99%	1 in 37600
Cohen syndrome (AR) NM_017890.4	VPS13B	Pan-ethnic	≤1 in 500	99%	Reduced
COL11A2-related conditions (AR) NM_080680.2	COL11A2 *	Pan-ethnic	≤1 in 500	99%	Reduced
COL17A1-related conditions (AR) NM_000494.3	COL17A1	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 8600
Combined oxidative phosphorylation deficiency 1 (AR) NM_024996.5	GFM1	Pan-ethnic	≤1 in 500	99%	Reduced
Combined oxidative phosphorylation deficiency 3 (AR) NM_001172696.1	TSMF *	Pan-ethnic	≤1 in 500	93%	Reduced
Combined pituitary hormone deficiency (LHX3-related) (AR) NM_014564.4	LHX3	Pan-ethnic	≤1 in 500	99%	Reduced
Combined pituitary hormone deficiency (POU1F1-related) (AR) NM_000306.3	POU1F1	Pan-ethnic	≤1 in 500	99%	Reduced
Combined pituitary hormone deficiency (PROT1-related) (AR) NM_006261.4	PROT1	Pan-ethnic	1 in 45	98%	1 in 2200
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency (AR) NM_000198.3	HSD3B2	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (AR) NM_000500.7	CYP21A2 *	Pan-ethnic	1 in 61	92%	1 in 751
Congenital adrenal insufficiency (AR) NM_000781.2	CYP11A1	Pan-ethnic	≤1 in 500	99%	Reduced



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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
Congenital chronic diarrhea (DGAT1-related) (AR) NM_012079.5	DGAT1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation (SLC35A3-related) (AR) NM_012243.2	SLC35A3	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation type Ia (AR) NM_000303.2	PMM2	Pan-ethnic	1 in 190	99%	1 in 18900
Congenital disorder of glycosylation type Ib (AR) NM_002435.2	MPI	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation type Ic (AR) NM_013339.3	ALG6	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation type Ik (AR) NM_019109.4	ALG1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital disorder of glycosylation type Iv (AR) NM_018297.3	NGLY1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital dyserythropoietic anemia type II (AR) NM_006363.4	SEC23B	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital hydrocephalus-1 (AR) NM_001080414.3	CCDC88C	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital hypothyroidism (TSHB-related) (AR) NM_000549.4	TSHB	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital insensitivity to pain with anhidrosis (AR) NM_001012331.1	NTRK1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital myasthenic syndrome (CHAT-related) (AR) NM_020549.4	CHAT	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital myasthenic syndrome (CHRNE-related) (AR) NM_000080.3	CHRNE	Pan-ethnic	1 in 200	99%	1 in 19900
Congenital nephrotic syndrome type 1 (AR) NM_004646.3	NPHS1	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital nephrotic syndrome type 2 (AR) NM_014625.3	NPHS2	Pan-ethnic	≤1 in 500	99%	Reduced
Congenital secretory chloride diarrhea (AR) NM_000111.2	SLC26A3	Pan-ethnic	≤1 in 500	99%	Reduced
Corneal dystrophy and perceptive deafness (AR) NM_032034.3	SLC4A11	Pan-ethnic	≤1 in 500	99%	Reduced
CRB1-related conditions (AR) NM_201253.2	CRB1	Pan-ethnic	1 in 112	99%	1 in 11100
CTSC-related conditions (AR) NM_001814.5	CTSC	Pan-ethnic	1 in 250	99%	1 in 24900
CYP1B1-related conditions (AR) NM_000104.3	CYP1B1	Pan-ethnic	1 in 79	99%	1 in 7800
CYP7B1-related conditions (AR) NM_004820.3	CYP7B1	Pan-ethnic	≤1 in 500	99%	Reduced
CYP11B1-related conditions (AR) NM_000497.3	CYP11B1	Pan-ethnic	1 in 194	99%	1 in 19300
CYP17A1-related conditions (AR) NM_000102.3	CYP17A1	Pan-ethnic	≤1 in 500	99%	Reduced
Cystinosis (AR) NM_004937.2	CTNS	Pan-ethnic	1 in 158	99%	1 in 15700
Cytochrome P450 oxidoreductase deficiency (AR) NM_000941.2	POR	Pan-ethnic	1 in 158	99%	1 in 15700
Desbuquois dysplasia type 1 (AR) NM_138793.3	CANT1	Pan-ethnic	≤1 in 500	99%	Reduced
Developmental and epileptic encephalopathy (CAD-related) (AR) NM_004341.4	CAD	Pan-ethnic	≤1 in 500	99%	Reduced
DGUOK-related conditions (AR) NM_080916.2	DGUOK	Pan-ethnic	≤1 in 500	99%	Reduced
DHDDS-related conditions (AR) NM_024887.3	DHDDS	Pan-ethnic	≤1 in 500	99%	Reduced
Dihydrolipoamide dehydrogenase deficiency (AR) NM_000108.4	DLD	Pan-ethnic	≤1 in 500	99%	Reduced



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Distal renal tubular acidosis with deafness (ATP6V1B1-related) (AR) NM_001692.3	ATP6V1B1	Pan-ethnic	≤1 in 500	99%	Reduced
DOK7-related conditions (AR) NM_173660.4	DOK7	Pan-ethnic	1 in 115	99%	1 in 11400
Donnai-Barrow syndrome (AR) NM_004525.2	LRP2	Pan-ethnic	≤1 in 500	99%	Reduced
Dubin-Johnson syndrome (AR) NM_000392.4	ABCC2 *	Pan-ethnic	≤1 in 500	99%	Reduced
DUOX2-related conditions (AR) NM_014080.4	DUOX2 *	Pan-ethnic	1 in 58	91%	1 in 634
DYNC2H1-related conditions (AR) NM_001080463.1	DYNC2H1	Pan-ethnic	1 in 224	99%	1 in 22300
DYSF-related conditions (AR) NM_003494.3	DYSF	Pan-ethnic	1 in 311	99%	1 in 31000
Dyskeratosis congenita spectrum disorders (RTEL1-related) (AR) NM_001283009.1	RTEL1	Pan-ethnic	≤1 in 500	99%	Reduced
Dyskeratosis congenita spectrum disorders (TERT-related) (AR) NM_198253.2	TERT	Pan-ethnic	≤1 in 500	99%	Reduced
Dystrophic epidermolysis bullosa (AR) NM_000094.3	COL7A1	Pan-ethnic	1 in 370	97%	1 in 12300
Ehlers-Danlos syndrome, dermatosparaxis type (AR) NM_014244.4	ADAMTS2	Pan-ethnic	≤1 in 500	99%	Reduced
Ehlers-Danlos syndrome, kyphoscoliotic type (AR) NM_000302.3	PLOD1	Pan-ethnic	1 in 150	99%	1 in 14900
Ellis-van Creveld syndrome (EVC-related) (AR) NM_153717.2	EVC	Pan-ethnic	1 in 220	99%	1 in 21900
Epidermolysis bullosa with pyloric atresia (ITGB4-related) (AR) NM_001005731.2	ITGB4	Pan-ethnic	1 in 393	99%	1 in 39200
Epimerase deficiency galactosemia (AR) NM_000403.3	GALE *	Pan-ethnic	≤1 in 500	99%	Reduced
ERCC2-related conditions (AR) NM_000400.3	ERCC2	Pan-ethnic	≤1 in 500	99%	Reduced
Ethylmalonic encephalopathy (AR) NM_014297.3	ETHE1	Pan-ethnic	≤1 in 500	99%	Reduced
EVC2-related conditions (AR) NM_147127.4	EVC2	Pan-ethnic	1 in 199	99%	1 in 19800
Familial chylomicronemia syndrome (AR) NM_000237.2	LPL	Pan-ethnic	≤1 in 500	99%	Reduced
Familial dysautonomia (AR) NM_003640.3	ELP1	Pan-ethnic	≤1 in 500	99%	Reduced
Familial hemophagocytic lymphohistiocytosis type 2 (AR) NM_001083116.1	PRF1	Pan-ethnic	1 in 177	99%	1 in 17600
Familial hemophagocytic lymphohistiocytosis type 3 (AR) NM_199242.2	UNC13D	Pan-ethnic	1 in 177	93%	1 in 2515
Familial hemophagocytic lymphohistiocytosis type 4 (AR) NM_003764.3	STX11	Pan-ethnic	1 in 224	99%	1 in 22300
Familial hemophagocytic lymphohistiocytosis type 5 (AR) NM_006949.3	STXBP2	Pan-ethnic	1 in 224	99%	1 in 22300
Familial hypercholesterolemia (LDLR-related) (AD) NM_000527.4	LDLR	Pan-ethnic	1 in 250	99%	1 in 24900
Familial hypercholesterolemia (LDLRAP1-related) (AR) NM_015627.2	LDLRAP1	Pan-ethnic	≤1 in 500	99%	Reduced
Fanconi anemia type A (AR) NM_000135.2	FANCA	Pan-ethnic	1 in 345	99%	1 in 34400
Fanconi anemia type C (AR) NM_000136.2	FANCC	Pan-ethnic	1 in 417	99%	1 in 41600
Fanconi anemia type D2 (AR) NM_033084.3	FANCD2 *	Pan-ethnic	≤1 in 500	94%	Reduced



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Fanconi anemia type E (AR) NM_021922.2	FANCE	Pan-ethnic	≤1 in 500	99%	Reduced
Fanconi anemia type G (AR) NM_004629.1	FANCG	Pan-ethnic	≤1 in 500	99%	Reduced
Fanconi anemia type I (AR) NM_001113378.1	FANCI	Pan-ethnic	≤1 in 500	99%	Reduced
Fanconi anemia type L (AR) NM_018062.3	FANCL *	Pan-ethnic	≤1 in 500	99%	Reduced
FH-related conditions (AR) NM_000143.3	FH *	Pan-ethnic	≤1 in 500	99%	Reduced
FKBP10-related conditions (AR) NM_021939.3	FKBP10	Pan-ethnic	≤1 in 500	99%	Reduced
Foveal hypoplasia (SLC38A8-related) (AR) NM_001080442.2	SLC38A8	Pan-ethnic	≤1 in 500	99%	Reduced
FOXN1-related conditions (AR) NM_003593.2	FOXN1	Pan-ethnic	≤1 in 500	99%	Reduced
Fraser syndrome (FRAS1-related) (AR) NM_025074.6	FRAS1	Pan-ethnic	1 in 316	99%	1 in 31500
Fraser syndrome (FREM2-related) (AR) NM_207361.5	FREM2	Pan-ethnic	≤1 in 500	99%	Reduced
Fraser syndrome (GRIP1-related) (AR) NM_021150.3	GRIP1	Pan-ethnic	1 in 447	99%	1 in 44600
Fructose-1,6-bisphosphatase deficiency (AR) NM_000507.3	FBP1	Pan-ethnic	≤1 in 500	99%	Reduced
Fucosidosis (AR) NM_000147.4	FUCA1	Pan-ethnic	≤1 in 500	99%	Reduced
Galactokinase deficiency galactosemia (AR) NM_000154.1	GALK1	Pan-ethnic	1 in 122	99%	1 in 12100
Galactosemia (GALT-related) (AR) NM_000155.3	GALT	Pan-ethnic	1 in 100	99%	1 in 9900
Galactosialidosis (AR) NM_000308.3	CTSA	Pan-ethnic	≤1 in 500	99%	Reduced
GATM-related conditions (AR) NM_001482.2	GATM	Pan-ethnic	≤1 in 500	99%	Reduced
GBA-related conditions including 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
GBE1-related conditions (AR) NM_000158.3	GBE1	Pan-ethnic	1 in 387	99%	1 in 38600
GCH1-related conditions (AR) NM_000161.2	GCH1	Pan-ethnic	≤1 in 500	99%	Reduced
GDF5-related conditions (AR) NM_000557.4	GDF5	Pan-ethnic	≤1 in 500	99%	Reduced
Geroderma osteodysplastica (AR) NM_152281.2	GORAB	Pan-ethnic	≤1 in 500	99%	Reduced
GHR-related conditions (AR) NM_000163.4	GHR *	Pan-ethnic	≤1 in 500	98%	Reduced
Gitelman syndrome (AR) NM_000339.2	SLC12A3	Pan-ethnic	1 in 100	99%	1 in 9900
GJB2-related conditions (AR) NM_004004.5	GJB2	Pan-ethnic	1 in 50	99%	1 in 4900
GLB1-related conditions (AR) NM_000404.2	GLB1	Pan-ethnic	1 in 158	99%	1 in 15700
GLE1-related conditions (AR) NM_001003722.1	GLE1	Pan-ethnic	≤1 in 500	99%	Reduced
Glutaric acidemia type I (AR) NM_000159.3	GCDH	Pan-ethnic	1 in 87	99%	1 in 8600
Glutaric acidemia type IIA (AR) NM_000126.3	ETFA	Pan-ethnic	≤1 in 500	99%	Reduced
Glutaric acidemia type IIB (AR) NM_001985.2	ETFB	Pan-ethnic	≤1 in 500	99%	Reduced
Glutaric acidemia type IIC (AR) NM_004453.3	ETFDH	Pan-ethnic	1 in 250	99%	1 in 24900



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DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
Glutathione synthetase deficiency (AR) NM_000178.2	GSS	Pan-ethnic	≤1 in 500	99%	Reduced
Glycine encephalopathy (AMT-related) (AR) NM_000481.3	AMT	Pan-ethnic	1 in 325	99%	1 in 32400
Glycine encephalopathy (GLDC-related) (AR) NM_000170.2	GLDC	Pan-ethnic	1 in 165	99%	1 in 16400
Glycogen storage disease type Ia (AR) NM_000151.3	G6PC	Pan-ethnic	1 in 177	95%	1 in 3520
Glycogen storage disease type II (Pompe disease) (AR) NM_000152.3	GAA	Pan-ethnic	1 in 100	99%	1 in 9900
Glycogen storage disease type III (AR) NM_000642.2	AGL	Pan-ethnic	1 in 159	95%	1 in 3160
Glycogen storage disease type IXb (AR) NM_000293.2	PHKB	Pan-ethnic	≤1 in 500	99%	Reduced
Glycogen storage disease type IXc (AR) NM_000294.2	PHKG2	Pan-ethnic	≤1 in 500	99%	Reduced
Glycogen storage disease type V (AR) NM_005609.3	PYGM	Pan-ethnic	1 in 171	99%	1 in 17000
Glycogen storage disease type VII (AR) NM_000289.5	PFKM	Pan-ethnic	≤1 in 500	99%	Reduced
GM3 synthase deficiency (AR) NM_003896.3	ST3GAL5	Pan-ethnic	≤1 in 500	99%	Reduced
GNE-related conditions (AR) NM_001128227.2	GNE *	Pan-ethnic	1 in 179	99%	1 in 17800
GNPTAB-related conditions (AR) NM_024312.4	GNPTAB	Pan-ethnic	1 in 200	99%	1 in 19900
Guanidinoacetate methyltransferase deficiency (AR) NM_000156.5	GAMT	Pan-ethnic	≤1 in 500	99%	Reduced
GUCY2D-related conditions (AR) NM_000180.3	GUCY2D	Pan-ethnic	1 in 204	99%	1 in 20300
Gyrate atrophy of the choroid and retina (AR) NM_000274.3	OAT *	Pan-ethnic	≤1 in 500	99%	Reduced
HADHA-related conditions (AR) NM_000182.4	HADHA	Pan-ethnic	1 in 350	99%	1 in 34900
HBB-related hemoglobinopathies (AR) NM_000518.4	HBB	Pan-ethnic	1 in 49	99%	1 in 4800
Heme oxygenase 1 deficiency (AR) NM_002133.2	HMOX1	Pan-ethnic	≤1 in 500	99%	Reduced
Hemolytic anemia, CD59-mediated (AR) NM_203330.2	CD59	Pan-ethnic	≤1 in 500	99%	Reduced
Hereditary fructose intolerance (AR) NM_000035.3	ALDOB	Pan-ethnic	1 in 122	99%	1 in 12100
Hereditary hemochromatosis type 2 (HAMP-related) (AR) NM_021175.2	HAMP	Pan-ethnic	≤1 in 500	99%	Reduced
Hereditary hemochromatosis type 2 (HJV-related) (AR) NM_213653.3	HJV	Pan-ethnic	≤1 in 500	99%	Reduced
Hereditary hemochromatosis type 3 (AR) NM_003227.3	TFR2	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 1 (AR) NM_000195.4	HPS1	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 3 (AR) NM_032383.4	HPS3	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 4 (AR) NM_022081.5	HPS4	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 5 (AR) NM_181507.1	HPS5	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 6 (AR) NM_024747.5	HPS6	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 8 (AR) NM_212550.4	BLOC1S3	Pan-ethnic	≤1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 9 (AR) NM_012388.3	BLOC1S6	Pan-ethnic	≤1 in 500	99%	Reduced



DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
HGSNAT-related conditions (AR) NM_152419.2	HGSNAT	Pan-ethnic	≤1 in 500	99%	Reduced
Holocarboxylase synthetase deficiency (AR) NM_000411.6	HLCS	Pan-ethnic	1 in 224	99%	1 in 22300
Homocystinuria due to cobalamin E deficiency (AR) NM_002454.2	MTRR	Pan-ethnic	≤1 in 500	99%	Reduced
Homocystinuria due to cobalamin G deficiency (AR) NM_000254.2	MTR	Pan-ethnic	≤1 in 500	99%	Reduced
Homocystinuria due to cystathionine beta-synthase deficiency (AR) NM_000071.2	CBS	Pan-ethnic	1 in 224	99%	1 in 22300
Homocystinuria due to MTHFR deficiency (AR) NM_005957.4	MTHFR *	Pan-ethnic	≤1 in 500	99%	Reduced
HSD17B4-related conditions (AR) NM_000414.3	HSD17B4	Pan-ethnic	1 in 158	99%	1 in 15700
Hydrolethalus syndrome type 1 (AR) NM_145014.2	HYLS1	Pan-ethnic	≤1 in 500	99%	Reduced
Hyper-IgM immunodeficiency (CD40-related) (AR) NM_001250.5	CD40	Pan-ethnic	≤1 in 500	99%	Reduced
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (AR) NM_014252.3	SLC25A15	Pan-ethnic	≤1 in 500	99%	Reduced
Hyperphosphatemic familial tumoral calcinosis (GALNT3-related) (AR) NM_004482.3	GALNT3	Pan-ethnic	≤1 in 500	99%	Reduced
Hypomyelinating leukodystrophy-12 (AR) NM_021729.5	VPS11	Pan-ethnic	≤1 in 500	99%	Reduced
Hypophosphatasia (AR) NM_000478.5	ALPL	Pan-ethnic	1 in 150	95%	1 in 2980
Ichthyosis prematurity syndrome (AR) NM_005094.3	SLC27A4	Pan-ethnic	≤1 in 500	99%	Reduced
IGHMBP2-related conditions (AR) NM_002180.2	IGHMBP2	Pan-ethnic	≤1 in 500	99%	Reduced
IKBKB-related conditions (AR) NM_001556.2	IKBKB	Pan-ethnic	≤1 in 500	99%	Reduced
Imerslund-Gräsbeck syndrome (AR) NM_030943.3	AMN *	Pan-ethnic	≤1 in 500	99%	Reduced
Immunodeficiency-centromeric instability-facial anomalies syndrome 1 (AR) NM_006892.3	DNMT3B	Pan-ethnic	≤1 in 500	99%	Reduced
Immunodeficiency-centromeric instability-facial anomalies syndrome 2 (AR) NM_014797.2	ZBTB24	Pan-ethnic	≤1 in 500	99%	Reduced
Isolated ectopia lentis (AR) NM_019032.5	ADAMTSL4	Pan-ethnic	≤1 in 500	99%	Reduced
Isovaleric acidemia (AR) NM_002225.3	IVD	Pan-ethnic	1 in 250	99%	1 in 24900
ITGB3-related conditions (AR) NM_000212.2	ITGB3	Pan-ethnic	≤1 in 500	99%	Reduced
Johanson-Blizzard syndrome (AR) NM_174916.2	UBR1	Pan-ethnic	1 in 250	99%	1 in 24900
Joubert syndrome and related disorders (MKS1-related) (AR) NM_017777.3	MKS1	Pan-ethnic	1 in 260	95%	1 in 5180
Joubert syndrome and related disorders (RPGRIPL-related) (AR) NM_015272.2	RPGRIPL	Pan-ethnic	1 in 259	95%	1 in 5160
Joubert syndrome and related disorders (TMEM216-related) (AR) NM_001173990.2	TMEM216	Pan-ethnic	≤1 in 500	99%	Reduced
Junctional epidermolysis bullosa (LAMC2-related) (AR) NM_005562.2	LAMC2	Pan-ethnic	≤1 in 500	99%	Reduced



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Junctional epidermolysis bullosa with pyloric atresia (ITGA6-related) (AR) NM_000210.3	ITGA6	Pan-ethnic	≤1 in 500	99%	Reduced
KCNJ11-related conditions (AR) NM_000525.3	KCNJ11	Pan-ethnic	≤1 in 500	99%	Reduced
Krabbe disease (AR) NM_000153.3	GALC *	Pan-ethnic	1 in 158	99%	1 in 15700
LAMA2-related muscular dystrophy (AR) NM_000426.3	LAMA2	Pan-ethnic	1 in 87	99%	1 in 8600
LAMA3-related conditions (AR) NM_000227.4	LAMA3	Pan-ethnic	≤1 in 500	99%	Reduced
LAMB3-related conditions (AR) NM_000228.2	LAMB3	Pan-ethnic	1 in 317	99%	1 in 31600
Leber congenital amaurosis 5 (AR) NM_181714.3	LCA5	Pan-ethnic	≤1 in 500	97%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B1-related) (AR) NM_001414.3	EIF2B1	Pan-ethnic	≤1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B2-related) (AR) NM_014239.3	EIF2B2	Pan-ethnic	≤1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B3-related) (AR) NM_020365.4	EIF2B3	Pan-ethnic	≤1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B4-related) (AR) NM_015636.3	EIF2B4	Pan-ethnic	≤1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B5-related) (AR) NM_003907.2	EIF2B5	Pan-ethnic	≤1 in 500	99%	Reduced
LIG4 syndrome (AR) NM_002312.3	LIG4	Pan-ethnic	≤1 in 500	99%	Reduced
Limb-girdle muscular dystrophy (CAPN3-related) (AR) NM_000070.2	CAPN3	Pan-ethnic	1 in 134	99%	1 in 13300
Limb-girdle muscular dystrophy type 2C (AR) NM_000231.2	SGCG	Pan-ethnic	≤1 in 500	99%	Reduced
Limb-girdle muscular dystrophy type 2D (AR) NM_000023.2	SGCA	Pan-ethnic	≤1 in 500	99%	Reduced
Limb-girdle muscular dystrophy type 2E (AR) NM_000232.4	SGCB	Pan-ethnic	≤1 in 500	92%	Reduced
Limb-girdle muscular dystrophy type 2F (AR) NM_000337.5	SGCD	Pan-ethnic	≤1 in 500	99%	Reduced
Lipoid congenital adrenal hyperplasia (AR) NM_000349.2	STAR	Pan-ethnic	≤1 in 500	99%	Reduced
LRAT-related conditions (AR) NM_004744.4	LRAT	Pan-ethnic	1 in 296	99%	1 in 29500
Lysinuric protein intolerance (AR) NM_001126106.2	SLC7A7	Pan-ethnic	≤1 in 500	99%	Reduced
Lysosomal acid lipase deficiency (AR) NM_000235.3	LIPA	Pan-ethnic	1 in 359	94%	1 in 5967
Major histocompatibility complex class II deficiency (CIITA-related) (AR) NM_000246.3	CIITA	Pan-ethnic	≤1 in 500	99%	Reduced
Malonyl-CoA decarboxylase deficiency (AR) NM_012213.2	MLYCD	Pan-ethnic	≤1 in 500	99%	Reduced
Maple syrup urine disease type 1A (AR) NM_000709.3	BCKDHA	Pan-ethnic	1 in 373	99%	1 in 37200
Maple syrup urine disease type 1B (AR) NM_183050.2	BCKDHB	Pan-ethnic	1 in 346	99%	1 in 34500
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	Pan-ethnic	1 in 66	99%	1 in 6500

DISORDER (INHERITANCE)	GENE	ETHNICITY	CARRIER FREQUENCY	DETECTION RATE	RISK TO BE A CARRIER AFTER NEGATIVE RESULT
Medium/short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (AR) NM_005327.4	HADH	Pan-ethnic	≤1 in 500	99%	Reduced
MEDNIK syndrome (AR) NM_001283.3	AP1S1	Pan-ethnic	≤1 in 500	99%	Reduced
Megalencephalic leukoencephalopathy with subcortical cysts 1 (AR) NM_015166.3	MLC1 *	Pan-ethnic	≤1 in 500	99%	Reduced
Metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration (AR) NM_152906.6	TANGO2	Pan-ethnic	≤1 in 500	99%	Reduced
Metachromatic leukodystrophy (ARSA-related) (AR) NM_000487.5	ARSA	Pan-ethnic	1 in 100	95%	1 in 1980
Methylmalonic acidemia (MCEE-related) (AR) NM_032601.3	MCEE	Pan-ethnic	≤1 in 500	99%	Reduced
Methylmalonic acidemia (MMAA-related) (AR) NM_172250.2	MMAA	Pan-ethnic	1 in 316	97%	1 in 10500
Methylmalonic acidemia (MMAB-related) (AR) NM_052845.3	MMAB	Pan-ethnic	1 in 456	98%	1 in 22750
Methylmalonic acidemia (MUT-related) (AR) NM_000255.3	MUT	Pan-ethnic	1 in 204	96%	1 in 5075
MFSD8-related conditions (AR) NM_152778.2	MFSD8	Pan-ethnic	≤1 in 500	99%	Reduced
Microcephalic osteodysplastic primordial dwarfism type II (AR) NM_006031.5	PCNT	Pan-ethnic	≤1 in 500	99%	Reduced
Microcephaly, postnatal progressive, with seizures and brain atrophy (AR) NM_004268.4	MED17	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex I deficiency 1 (AR) NM_002495.3	NDUFS4	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex I deficiency 3 (AR) NM_024407.4	NDUFS7	Pan-ethnic	1 in 387	99%	1 in 38600
Mitochondrial complex I deficiency 4 (AR) NM_007103.3	NDUFV1	Pan-ethnic	1 in 387	99%	1 in 38600
Mitochondrial complex I deficiency 9 (AR) NM_004553.4	NDUFS6	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex I deficiency 10 (AR) NM_174889.4	NDUFAF2	Pan-ethnic	1 in 387	99%	1 in 38600
Mitochondrial complex I deficiency 16 (AR) NM_024120.4	NDUFAF5	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex I deficiency 19 (AR) NM_017547.3	FOXRED1	Pan-ethnic	1 in 376	99%	1 in 37500
Mitochondrial complex I deficiency 20/ACAD9 deficiency (AR) NM_014049.4	ACAD9	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex IV deficiency 6 (AR) NM_004376.6	COX15	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial complex IV deficiency 12 (AR) NM_001171155.1	PET100	Pan-ethnic	1 in 387	99%	1 in 38600
Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (AR) NM_133259.3	LRPPRC	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial DNA depletion syndrome-2 (AR) NM_004614.4	TK2	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial neurogastrointestinal encephalomyopathy (AR) NM_001953.4	TYMP	Pan-ethnic	≤1 in 500	99%	Reduced
Mitochondrial trifunctional protein deficiency (HADHB-related) (AR) NM_000183.2	HADHB	Pan-ethnic	≤1 in 500	99%	Reduced
MKKS-related conditions (AR) NM_018848.3	MKKS	Pan-ethnic	≤1 in 500	99%	Reduced



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Molybdenum cofactor deficiency (MOCS1-related) (AR) NM_001358530.2	MOCS1	Pan-ethnic	1 in 226	99%	1 in 22500
Molybdenum cofactor deficiency (MOCS2-related) (AR) NM_004531.4	MOCS2B	Pan-ethnic	≤1 in 500	99%	Reduced
Molybdenum cofactor deficiency (MOCS2-related) (AR) NM_176806.3	MOCS2A	Pan-ethnic	≤1 in 500	99%	Reduced
MPL-related conditions (AR) NM_005373.2	MPL	Pan-ethnic	≤1 in 500	99%	Reduced
MPV17-related conditions (AR) NM_002437.4	MPV17	Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolipidosis type III gamma (AR) NM_032520.4	GNPTG	Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolipidosis type IV (AR) NM_020533.2	MCOLN1	Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolysaccharidosis type I (AR) NM_000203.4	IDUA	Pan-ethnic	1 in 148	97%	1 in 4900
Mucopolysaccharidosis type IIIA (AR) NM_000199.3	SGSH	Pan-ethnic	1 in 215	99%	1 in 21400
Mucopolysaccharidosis type IIIB (AR) NM_000263.3	NAGLU	Pan-ethnic	1 in 224	99%	1 in 22300
Mucopolysaccharidosis type IIID (AR) NM_002076.3	GNS	Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolysaccharidosis type IVA (AR) NM_000512.4	GALNS	Pan-ethnic	1 in 224	99%	1 in 22300
Mucopolysaccharidosis type IX (AR) NM_153281.1	HYAL1	Pan-ethnic	≤1 in 500	99%	Reduced
Mucopolysaccharidosis type VI (AR) NM_000046.3	ARSB	Pan-ethnic	1 in 250	99%	1 in 24900
Mucopolysaccharidosis type VII (AR) NM_000181.3	GUSB	Pan-ethnic	1 in 250	99%	1 in 24900
Mulibrey nanism (AR) NM_015294.4	TRIM37	Pan-ethnic	≤1 in 500	99%	Reduced
Multiple pterygium syndrome (AR) NM_005199.4	CHRNA3	Pan-ethnic	≤1 in 500	99%	Reduced
Multiple sulfatase deficiency (AR) NM_182760.3	SUMF1	Pan-ethnic	≤1 in 500	99%	Reduced
Muscular dystrophy-dystroglycanopathy (FKRP-related) (AR) NM_024301.4	FKRP	Pan-ethnic	1 in 158	99%	1 in 15700
Muscular dystrophy-dystroglycanopathy (FKTN-related) (AR) NM_001079802.1	FKTN	Pan-ethnic	≤1 in 500	99%	Reduced
Muscular dystrophy-dystroglycanopathy (LARGE1-related) (AR) NM_004737.4	LARGE1	Pan-ethnic	≤1 in 500	99%	Reduced
Muscular dystrophy-dystroglycanopathy (POMT1-related) (AR) NM_007171.3	POMT1	Pan-ethnic	1 in 268	99%	1 in 26700
Muscular dystrophy-dystroglycanopathy (POMT2-related) (AR) NM_013382.5	POMT2	Pan-ethnic	1 in 371	99%	1 in 37000
Muscular dystrophy-dystroglycanopathy (RXYL1-related) (AR) NM_014254.2	RXYLT1	Pan-ethnic	≤1 in 500	99%	Reduced
MUSK-related conditions (AR) NM_005592.3	MUSK	Pan-ethnic	1 in 447	99%	1 in 44600
MVK-related conditions (AR) NM_000431.3	MVK	Pan-ethnic	≤1 in 500	99%	Reduced
MYO7A-related conditions (AR) NM_000260.3	MYO7A	Pan-ethnic	1 in 200	95%	1 in 3980
Myopathy, lactic acidosis, and sideroblastic anemia 1 (AR) NM_025215.5	PUS1	Pan-ethnic	≤1 in 500	99%	Reduced



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Myotonia congenita (AR) NM_000083.2	CLCN1	Pan-ethnic	1 in 112	99%	1 in 11100
N-acetylglutamate synthase deficiency (AR) NM_153006.2	NAGS	Pan-ethnic	≤1 in 500	99%	Reduced
Nemaline myopathy 2 (AR) NM_001271208.1	NEB *	Pan-ethnic	1 in 158	95%	1 in 3140
Nephrogenic diabetes insipidus (AQP2-related) (AR) NM_000486.5	AQP2	Pan-ethnic	≤1 in 500	99%	Reduced
Nephronophthisis (INVS-related) (AR) NM_014425.3	INVS	Pan-ethnic	≤1 in 500	99%	Reduced
Nephronophthisis (NPHP1-related) (AR) NM_000272.3	NPHP1	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 1 (AR) NM_000310.3	PPT1	Pan-ethnic	1 in 199	98%	1 in 9900
Neuronal ceroid lipofuscinosis type 2 (AR) NM_000391.3	TPP1	Pan-ethnic	1 in 250	97%	1 in 8300
Neuronal ceroid lipofuscinosis type 5 (AR) NM_006493.2	CLN5	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 6 (AR) NM_017882.2	CLN6	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 8 (AR) NM_018941.3	CLN8	Pan-ethnic	≤1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 10 (AR) NM_001909.4	CTSD	Pan-ethnic	≤1 in 500	99%	Reduced
Niemann-Pick disease type C (NPC1-related) (AR) NM_000271.4	NPC1	Pan-ethnic	1 in 183	99%	1 in 18200
Niemann-Pick disease type C (NPC2-related) (AR) NM_006432.3	NPC2	Pan-ethnic	≤1 in 500	99%	Reduced
Niemann-Pick disease types A and B (AR) NM_000543.4	SMPD1	Pan-ethnic	1 in 250	95%	1 in 4980
Nijmegen breakage syndrome (AR) NM_002485.4	NBN	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic deafness (LOXHD1-related) (AR) NM_144612.6	LOXHD1	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic deafness (MYO15A-related) (AR) NM_016239.3	MYO15A	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic deafness (OTOA-related) (AR) NM_144672.3	OTOA *	Pan-ethnic	≤1 in 500	88%	Reduced
Nonsyndromic deafness (SYNE4-related) (AR) NM_001039876.2	SYNE4	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic deafness (TMC1-related) (AR) NM_138691.2	TMC1	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic deafness (TMPRSS3-related) (AR) NM_024022.2	TMPRSS3	Pan-ethnic	≤1 in 500	99%	Reduced
Nonsyndromic intellectual disability (CC2D1A-related) (AR) NM_017721.5	CC2D1A	Pan-ethnic	≤1 in 500	99%	Reduced
NR2E3-related conditions (AR) NM_014249.3	NR2E3	Pan-ethnic	≤1 in 500	99%	Reduced
NSMCE3 deficiency (AR) NM_138704.3	NSMCE3	Pan-ethnic	≤1 in 500	99%	Reduced
Oculocutaneous albinism type 2 (AR) NM_000275.2	OCA2	Pan-ethnic	1 in 95	99%	1 in 9400
Oculocutaneous albinism type 3 (AR) NM_000550.2	TYRP1	Pan-ethnic	≤1 in 500	99%	Reduced
Oculocutaneous albinism type 4 (AR) NM_016180.4	SLC45A2	Pan-ethnic	1 in 158	99%	1 in 15700
Oculocutaneous albinism types 1A and 1B (AR) NM_000372.4	TYR *	Pan-ethnic	1 in 100	97%	1 in 3300
OPA3-related conditions (AR) NM_025136.3	OPA3	Pan-ethnic	≤1 in 500	99%	Reduced
Osteogenesis imperfecta (BMP1-related) (AR) NM_006129.4	BMP1	Pan-ethnic	≤1 in 500	99%	Reduced



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Osteogenesis imperfecta (CRTAP-related) (AR) NM_006371.4	CRTAP	Pan-ethnic	≤1 in 500	99%	Reduced
Osteogenesis imperfecta (P3H1-related) (AR) NM_022356.3	P3H1	Pan-ethnic	≤1 in 500	99%	Reduced
Osteopetrosis (TCIRG1-related) (AR) NM_006019.3	TCIRG1	Pan-ethnic	1 in 317	99%	1 in 31600
OSTM1 deficiency associated osteopetrosis (AR) NM_014028.3	OSTM1	Pan-ethnic	≤1 in 500	99%	Reduced
OTOF-related conditions (AR) NM_194248.2	OTOF	Pan-ethnic	≤1 in 500	99%	Reduced
Pantothenate kinase-associated neurodegeneration (AR) NM_153638.2	PANK2	Pan-ethnic	1 in 289	99%	1 in 28800
Parkinson disease 15 (AR) NM_012179.3	FBXO7	Pan-ethnic	≤1 in 500	99%	Reduced
PCDH15-related conditions (AR) NM_033056.3	PCDH15	Pan-ethnic	1 in 400	99%	1 in 39900
PEX5-related conditions (AR) NM_001131025.1	PEX5	Pan-ethnic	≤1 in 500	99%	Reduced
PEX7-related conditions (AR) NM_000288.3	PEX7	Pan-ethnic	1 in 157	99%	1 in 15600
PGM3-congenital disorder of glycosylation (AR) NM_001199917.1	PGM3	Pan-ethnic	≤1 in 500	99%	Reduced
Phenylalanine hydroxylase deficiency (AR) NM_000277.1	PAH	Pan-ethnic	1 in 58	99%	1 in 5700
Phosphoglycerate dehydrogenase deficiency (AR) NM_006623.3	PHGDH	Pan-ethnic	≤1 in 500	99%	Reduced
PIGN-congenital disorder of glycosylation (AR) NM_176787.4	PIGN	Pan-ethnic	≤1 in 500	99%	Reduced
PJVK-related conditions (AR) NM_001042702.3	DFNB59	Pan-ethnic	≤1 in 500	99%	Reduced
PLA2G6-related conditions (AR) NM_003560.2	PLA2G6	Pan-ethnic	≤1 in 500	99%	Reduced
PLEKHG5-related conditions (AR) NM_020631.4	PLEKHG5	Pan-ethnic	≤1 in 500	99%	Reduced
POLG-related conditions (AR) NM_002693.2	POLG	Pan-ethnic	1 in 113	95%	1 in 2240
Polycystic kidney disease (PKHD1-related) (AR) NM_138694.3	PKHD1 *	Pan-ethnic	1 in 70	99%	1 in 6900
Polymicrogyria (ADGRG1-related) (AR) NM_005682.6	ADGRG1	Pan-ethnic	≤1 in 500	99%	Reduced
POMGNT1-related conditions (AR) NM_017739.3	POMGNT1	Pan-ethnic	≤1 in 500	99%	Reduced
Pontocerebellar hypoplasia (TSEN54-related) (AR) NM_207346.2	TSEN54	Pan-ethnic	≤1 in 500	99%	Reduced
Pontocerebellar hypoplasia type 1B (AR) NM_016042.3	EXOSC3	Pan-ethnic	≤1 in 500	99%	Reduced
Pontocerebellar hypoplasia type 2D (AR) NM_016955.3	SEPSECS	Pan-ethnic	≤1 in 500	99%	Reduced
Pontocerebellar hypoplasia type 6 (AR) NM_020320.3	RARS2	Pan-ethnic	≤1 in 500	99%	Reduced
Primary carnitine deficiency (AR) NM_003060.3	SLC22A5	Pan-ethnic	1 in 71	99%	1 in 7000
Primary ciliary dyskinesia (CCDC39-related) (AR) NM_181426.1	CCDC39	Pan-ethnic	1 in 211	99%	1 in 21000
Primary ciliary dyskinesia (CCDC103-related) (AR) NM_213607.2	CCDC103	Pan-ethnic	1 in 316	99%	1 in 31500
Primary ciliary dyskinesia (DNAH5-related) (AR) NM_001369.2	DNAH5	Pan-ethnic	1 in 109	99%	1 in 10800
Primary ciliary dyskinesia (DNAH11-related) (AR) NM_001277115.1	DNAH11	Pan-ethnic	1 in 211	99%	1 in 21000
Primary ciliary dyskinesia (DNAI1-related) (AR) NM_012144.3	DNAI1	Pan-ethnic	1 in 250	99%	1 in 24900



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Primary ciliary dyskinesia (DNAI2-related) (AR) NM_023036.4	DNAI2	Pan-ethnic	1 in 354	99%	1 in 35300
Primary hyperoxaluria type 1 (AR) NM_000030.2	AGXT	Pan-ethnic	1 in 135	99%	1 in 13400
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 35300
Primary microcephaly (MCPH1-related) (AR) NM_024596.4	MCPH1	Pan-ethnic	≤1 in 500	99%	Reduced
Progressive early-onset encephalopathy with brain atrophy and thin corpus callosum (PEBAT) (AR) NM_005993.4	TBCD	Pan-ethnic	≤1 in 500	99%	Reduced
Progressive pseudorheumatoid dysplasia (AR) NM_003880.3	WISP3	Pan-ethnic	≤1 in 500	99%	Reduced
Prolidase deficiency (AR) NM_000285.3	PEPD	Pan-ethnic	≤1 in 500	99%	Reduced
Propionic acidemia (PCCA-related) (AR) NM_000282.3	PCCA	Pan-ethnic	1 in 224	96%	1 in 5575
Propionic acidemia (PCCB-related) (AR) NM_000532.4	PCCB	Pan-ethnic	1 in 224	99%	1 in 22300
PSAP-related conditions (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 43700
Pyridoxal 5'-phosphate-dependent epilepsy (AR) NM_018129.3	PNPO	Pan-ethnic	≤1 in 500	99%	Reduced
Pyridoxine-dependent epilepsy (ALDH7A1-related) (AR) NM_001182.4	ALDH7A1	Pan-ethnic	1 in 127	99%	1 in 12600
Pyruvate carboxylase deficiency (AR) NM_000920.3	PC	Pan-ethnic	1 in 250	95%	1 in 4980
Pyruvate dehydrogenase complex deficiency (PDHB-related) (AR) NM_000925.3	PDHB	Pan-ethnic	≤1 in 500	99%	Reduced
RAPSN-related conditions (AR) NM_005055.4	RAPSN	Pan-ethnic	1 in 283	99%	1 in 28200
RDH12-related conditions (AR) NM_152443.2	RDH12	Pan-ethnic	1 in 460	99%	1 in 45900
Refsum disease (PHYH-related) (AR) NM_006214.3	PHYH	Pan-ethnic	≤1 in 500	99%	Reduced
Retinitis pigmentosa 25 (AR) NM_001142800.1	EYS *	Pan-ethnic	1 in 129	99%	1 in 12800
Retinitis pigmentosa 28 (AR) NM_001201543.1	FAM161A	Pan-ethnic	1 in 289	99%	1 in 28800
Retinitis pigmentosa 36 (AR) NM_001077620.2	PRCD	Pan-ethnic	1 in 296	99%	1 in 29500
Retinitis pigmentosa 62 (AR) NM_001242957.2	MAK	Pan-ethnic	1 in 274	99%	1 in 27300
Rhizomelic chondrodysplasia punctata type 2 (AR) NM_014236.3	GNPAT	Pan-ethnic	≤1 in 500	99%	Reduced
Rhizomelic chondrodysplasia punctata type 3 (AR) NM_003659.3	AGPS	Pan-ethnic	≤1 in 500	99%	Reduced
RLBP1-related conditions (AR) NM_000326.4	RLBP1	Pan-ethnic	1 in 296	99%	1 in 29500
Roberts syndrome (AR) NM_001017420.2	ESCO2	Pan-ethnic	≤1 in 500	99%	Reduced
RPE65-related conditions (AR) NM_000329.2	RPE65	Pan-ethnic	1 in 228	99%	1 in 22700
RYR1-related conditions (AR) NM_000540.2	RYR1	Pan-ethnic	≤1 in 500	99%	Reduced
SAMD9-related conditions (AR) NM_017654.3	SAMD9	Pan-ethnic	≤1 in 500	99%	Reduced



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Sandhoff disease (AR) NM_000521.3	HEXB	Pan-ethnic	1 in 180	99%	1 in 17900
Schimke immuno-osseous dysplasia (AR) NM_014140.3	SMARCAL1	Pan-ethnic	≤1 in 500	99%	Reduced
Seckel syndrome (CEP152-related) (AR) NM_014985.3	CEP152	Pan-ethnic	≤1 in 500	99%	Reduced
Sepiapterin reductase deficiency (AR) NM_003124.4	SPR	Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency due to CD3-delta deficiency (AR) NM_000732.4	CD3D	Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency due to CD3-epsilon deficiency (AR) NM_000733.3	CD3E	Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency due to CD45 deficiency (AR) NM_002838.4	PTPRC *	Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency due to DCLRE1C (Artemis) deficiency (AR) NM_001033855.2	DCLRE1C	Pan-ethnic	≤1 in 500	99%	Reduced
Severe combined immunodeficiency due to IL7R-alpha deficiency (AR) NM_002185.3	IL7R	Pan-ethnic	1 in 348	99%	1 in 34700
Severe combined immunodeficiency due to JAK3 deficiency (AR) NM_000215.3	JAK3	Pan-ethnic	1 in 455	99%	1 in 45400
Severe combined immunodeficiency due to RAG1 deficiency (AR) NM_000448.2	RAG1	Pan-ethnic	1 in 301	99%	1 in 30000
Severe combined immunodeficiency due to RAG2 deficiency (AR) NM_000536.3	RAG2	Pan-ethnic	≤1 in 500	99%	Reduced
Severe congenital neutropenia due to G6PC3 deficiency (AR) NM_138387.3	G6PC3	Pan-ethnic	≤1 in 500	99%	Reduced
Severe congenital neutropenia due to HAX1 deficiency (AR) NM_006118.3	HAX1	Pan-ethnic	≤1 in 500	99%	Reduced
Severe congenital neutropenia due to VPS45 deficiency (AR) NM_007259.4	VPS45	Pan-ethnic	≤1 in 500	99%	Reduced
Sialic acid storage diseases (AR) NM_012434.4	SLC17A5	Pan-ethnic	≤1 in 500	99%	Reduced
Sialidosis (AR) NM_000434.3	NEU1	Pan-ethnic	≤1 in 500	99%	Reduced
Sjögren-Larsson syndrome (AR) NM_000382.2	ALDH3A2	Pan-ethnic	≤1 in 500	99%	Reduced
SLC12A6-related conditions (AR) NM_133647.1	SLC12A6	Pan-ethnic	≤1 in 500	99%	Reduced
SLC26A4-related conditions (AR) NM_000441.1	SLC26A4	Pan-ethnic	1 in 80	99%	1 in 7900
SLC37A4-related conditions (AR) NM_001164277.1	SLC37A4	Pan-ethnic	1 in 354	95%	1 in 7060
Smith-Lemli-Opitz syndrome (AR) NM_001360.2	DHCR7	Pan-ethnic	1 in 71	99%	1 in 7000
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	Pan-ethnic	≤1 in 500	99%	Reduced
Spastic tetraplegia, thin corpus callosum, and progressive microcephaly (AR) NM_003038.4	SLC1A4	Pan-ethnic	≤1 in 500	99%	Reduced
SPG11-related conditions (AR) NM_025137.3	SPG11	Pan-ethnic	1 in 141	99%	1 in 14000





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Spinal muscular atrophy (AR) NM_000344.3 Carrier residual risks listed are for 2 copy SMN1 results. Carrier residual risk for >2 copies are 5- to 10-fold lower.	SMN1 *	African-American	1 in 59	83%	1 in 342
		Ashkenazi Jewish	1 in 62	94%	1 in 1017
		Asian	1 in 50	93%	1 in 701
		Caucasian	1 in 45	95%	1 in 880
		Hispanic	1 in 48	94%	1 in 784
		Pan-ethnic	1 in 49	94%	1 in 800
Spinocerebellar ataxia (ANO10-related) (AR) NM_018075.3	ANO10 *	Pan-ethnic	≤1 in 500	99%	Reduced
Spondylocostal dysostosis (DLL3-related) (AR) NM_016941.3	DLL3	Pan-ethnic	1 in 350	99%	1 in 34900
Spondylocostal dysostosis (MESP2-related) (AR) NM_001039958.1	MESP2	Pan-ethnic	1 in 224	99%	1 in 22300
Steel syndrome (AR) NM_032888.3	COL27A1	Pan-ethnic	≤1 in 500	99%	Reduced
Steroid 5-alpha-reductase deficiency (AR) NM_000348.3	SRD5A2	Pan-ethnic	≤1 in 500	99%	Reduced
Stüve-Wiedemann syndrome (AR) NM_002310.5	LIFR *	Pan-ethnic	≤1 in 500	99%	Reduced
Sulfite oxidase deficiency (AR) NM_000456.2	SUOX	Pan-ethnic	≤1 in 500	99%	Reduced
SURF1-related conditions (AR) NM_003172.3	SURF1	Pan-ethnic	1 in 128	99%	1 in 12700
Tay-Sachs disease (AR) NM_000520.4	HEXA	Pan-ethnic	1 in 250	99%	1 in 24900
TBCE-related conditions (AR) NM_003193.4	TBCE *	Pan-ethnic	≤1 in 500	99%	Reduced
Thiamine-responsive megaloblastic anemia (AR) NM_006996.2	SLC19A2	Pan-ethnic	≤1 in 500	99%	Reduced
Thyroid dysmorphogenesis (SLC5A5-related) (AR) NM_000453.2	SLC5A5	Pan-ethnic	≤1 in 500	99%	Reduced
Thyroid dysmorphogenesis (TG-related) (AR) NM_003235.4	TG *	Pan-ethnic	≤1 in 500	99%	Reduced
Thyroid dysmorphogenesis (TPO-related) (AR) NM_000547.5	TPO	Pan-ethnic	1 in 129	99%	1 in 12800
TMEM67-related conditions (AR) NM_153704.5	TMEM67	Pan-ethnic	1 in 316	99%	1 in 31500
Transcobalamin II deficiency (AR) NM_000355.3	TCN2	Pan-ethnic	≤1 in 500	99%	Reduced
Transient infantile liver failure (AR) NM_018006.4	TRMU	Pan-ethnic	≤1 in 500	99%	Reduced
TREX1-related conditions (AR) NM_033629.4	TREX1	Pan-ethnic	≤1 in 500	99%	Reduced
Trichohepatoenteric syndrome (SKIV2L-related) (AR) NM_006929.4	SKIV2L	Pan-ethnic	≤1 in 500	99%	Reduced
Trichohepatoenteric syndrome (TTC37-related) (AR) NM_014639.3	TTC37	Pan-ethnic	≤1 in 500	99%	Reduced
TRIM32-related conditions (AR) NM_012210.3	TRIM32	Pan-ethnic	1 in 408	99%	1 in 40700
Trimethylaminuria (AR) NM_006894.6	FMO3	Pan-ethnic	≤1 in 500	99%	Reduced
Triple A syndrome (AR) NM_015665.5	AAAS	Pan-ethnic	≤1 in 500	99%	Reduced
TSHR-related conditions (AR) NM_000369.2	TSHR	Pan-ethnic	1 in 158	99%	1 in 15700
TULP1-related conditions (AR) NM_003322.4	TULP1	Pan-ethnic	1 in 296	99%	1 in 29500
Tyrosine hydroxylase deficiency (AR) NM_199292.2	TH	Pan-ethnic	≤1 in 500	99%	Reduced
Tyrosinemia type I (AR) NM_000137.2	FAH *	Pan-ethnic	1 in 125	95%	1 in 2480
Tyrosinemia type II (AR) NM_000353.2	TAT	Pan-ethnic	1 in 250	99%	1 in 24900



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Tyrosinemia type III (AR) NM_002150.2	HPD	Pan-ethnic	≤1 in 500	99%	Reduced
USH1C-related conditions (AR) NM_005709.3	USH1C *	Pan-ethnic	1 in 353	90%	1 in 3521
USH2A-related conditions (AR) NM_206933.2	USH2A	Pan-ethnic	1 in 112	99%	1 in 11100
Very long-chain acyl-CoA dehydrogenase deficiency (AR) NM_000018.3	ACADVL	Pan-ethnic	1 in 100	99%	1 in 9900
Vici syndrome (AR) NM_020964.2	EPG5	Pan-ethnic	≤1 in 500	99%	Reduced
Vitamin D-dependent rickets type 1A (AR) NM_000785.3	CYP27B1	Pan-ethnic	≤1 in 500	99%	Reduced
Vitamin D-dependent rickets type 2A (AR) NM_001017535.1	VDR	Pan-ethnic	≤1 in 500	99%	Reduced
VPS53-related conditions (AR) NM_001128159.2	VPS53 *	Pan-ethnic	≤1 in 500	99%	Reduced
VRK1-related conditions (AR) NM_003384.2	VRK1	Pan-ethnic	≤1 in 500	99%	Reduced
VSX2-related conditions (AR) NM_182894.2	VSX2	Pan-ethnic	≤1 in 500	99%	Reduced
Warsaw syndrome (AR) NM_030653.3	DDX11 *	Pan-ethnic	≤1 in 500	15%	Reduced
Werner syndrome (AR) NM_000553.4	WRN *	Pan-ethnic	1 in 224	99%	1 in 22300
Wilson disease (AR) NM_000053.3	ATP7B	Pan-ethnic	1 in 90	98%	1 in 4450
WNT10A-related conditions (AR) NM_025216.2	WNT10A	Pan-ethnic	1 in 305	99%	1 in 30400
Wolcott-Rallison syndrome (AR) NM_004836.6	EIF2AK3	Pan-ethnic	≤1 in 500	99%	Reduced
Woodhouse-Sakati syndrome (AR) NM_025000.3	DCAF17	Pan-ethnic	≤1 in 500	99%	Reduced
Xeroderma pigmentosum complementation group A (AR) NM_000380.3	XPA	Pan-ethnic	≤1 in 500	99%	Reduced
Xeroderma pigmentosum complementation group C (AR) NM_004628.4	XPC	Pan-ethnic	≤1 in 500	99%	Reduced
Xeroderma pigmentosum, variant type (AR) NM_006502.2	POLH	Pan-ethnic	≤1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX1-related) (AR) NM_000466.2	PEX1 *	Pan-ethnic	1 in 144	99%	1 in 14300
Zellweger spectrum disorder (PEX2-related) (AR) NM_000318.2	PEX2	Pan-ethnic	≤1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX6-related) (AR) NM_000287.3	PEX6	Pan-ethnic	1 in 294	99%	1 in 29300
Zellweger spectrum disorder (PEX10-related) (AR) NM_153818.1	PEX10	Pan-ethnic	≤1 in 500	94%	Reduced
Zellweger spectrum disorder (PEX12-related) (AR) NM_000286.2	PEX12	Pan-ethnic	1 in 409	99%	1 in 40800
Zellweger spectrum disorder (PEX13-related) (AR) NM_002618.3	PEX13	Pan-ethnic	≤1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX16-related) (AR) NM_004813.2	PEX16	Pan-ethnic	≤1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX26-related) (AR) NM_017929.5	PEX26	Pan-ethnic	≤1 in 500	99%	Reduced