Sonic Genetics
Quality is in our DNA

Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
3-ketothiolase deficiency	ACAT1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
3-methylcrotonyl-CoA carboxylase 1 deficiency (3-MCC deficiency)	MCCC1	AR	General	1 in 95	98%	1 in 4,701	1 in 1,786,380
3-methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)	MCCC2	AR	General	1 in 95	98%	1 in 4,701	1 in 1,786,380
Abetalipoproteinaemia	MTTP	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Abetalipoproteinaemia	MTTP	AR	Ashkenazi Jewish	1 in 180	98%	1 in 8,951	1 in 6,444,720
Achondrogenesis, type 1B	SLC26A2	AR	General	1 in 158	90%	1 in 1,571	1 in 992,872
Achondrogenesis, type 1B	SLC26A2	AR	Finnish	1 in 50	90%	1 in 491	1 in 98,200
Achromatopsia	CNGB3	AR	General	1 in 87	99%	1 in 8,601	1 in 2,993,148
Achromatopsia	CNGB3	AR	Micronesian	1 in 2	99%	1 in 101	1 in 808
Acrodermatitis enteropathica	SLC39A4	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Acyl-CoA dehydrogenase-9 (ACAD9) deficiency	ACAD9	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Adenosine deaminase deficiency	ADA	AR	General	1 in 224	93%	1 in 3,187	1 in 2,855,552
Adrenoleucodystrophy, X-linked	ABCD1	XL	General	1 in 21,000	99%	1 in 2,099,901	1 in 8,399,804
Aicardi-Goutières syndrome	SAMHD1	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Alkaptonuria	HGD	AR	General	1 in 250	90%	1 in 2,491	1 in 2,491,000
Allan-Herndon-Dudley syndrome	SLC16A2	XL	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Alpers-Huttenlocher syndrome	POLG	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
Alpha thalassaemia	HBA1	AR	General	1 in 20	90%	1 in 191	1 in 15,280
Alpha thalassaemia	HBA1	AR	African/African American	1 in 3	90%	1 in 21	1 in 252
Alpha thalassaemia	HBA1	AR	Ashkenazi Jewish	1 in 13	90%	1 in 121	1 in 6,292
Alpha thalassaemia	HBA1	AR	East Asian	1 in 8	90%	1 in 71	1 in 2,272
Alpha thalassaemia	HBA1	AR	Middle-Eastern	1 in 3	90%	1 in 21	1 in 252
Alpha thalassaemia	HBA1	AR	South Asian/Indian	1 in 5	90%	1 in 41	1 in 820
Alpha thalassaemia	HBA2	AR	General	1 in 20	90%	1 in 191	1 in 15,280
Alpha thalassaemia	HBA2	AR	African/African American	1 in 3	90%	1 in 21	1 in 252
Alpha thalassaemia	HBA2	AR	Ashkenazi Jewish	1 in 13	90%	1 in 121	1 in 6,292
Alpha thalassaemia	HBA2	AR	East Asian	1 in 8	90%	1 in 71	1 in 2,272
Alpha thalassaemia	HBA2	AR	Middle-Eastern	1 in 3	90%	1 in 21	1 in 252
Alpha thalassaemia	HBA2	AR	South Asian/Indian	1 in 5	90%	1 in 41	1 in 820
Alpha thalassaemia X-linked intellectual disability syndrome	ATRX	XL	General	<1 in 250,000	99%	1 in 24,999,901	<1 in 10 million
Alpha-1 antitrypsin deficiency	SERPINA1	AR	General	1 in 33	95%	1 in 641	1 in 84,612
Alpha-1 antitrypsin deficiency	SERPINA1	AR	Caucasian/European	1 in 19	95%	1 in 361	1 in 27,436
Alpha-mannosidosis	MAN2B1	AR	General	1 in 354	99%	1 in 35,301	<1 in 10 million
Alpha-mannosidosis	MAN2B1	AR	Caucasian/European	1 in 274	99%	1 in 27,301	<1 in 10 million
Alport syndrome, COL4A3-related	COL4A3	AR	General	1 in 267	98%	1 in 13,301	<1 in 10 million
Alport syndrome, COL4A3-related	COL4A3	AR	Ashkenazi Jewish	1 in 188	98%	1 in 9,351	1 in 7,031,952
Alport syndrome, COL4A4-related	COL4A4	AR	General	1 in 267	98%	1 in 13,301	<1 in 10 million
Alport syndrome, COL4A5-related	COL4A5	XL	General	1 in 139	98%	1 in 6,901	1 in 27,604
Alstrom syndrome	ALMS1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Anauxetic dysplasia	RMRP	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Anauxetic dysplasia	RMRP	AR	Amish	1 in 16	99%	1 in 1,501	1 in 96,064
Anauxetic dysplasia	RMRP	AR	Finnish	1 in 76	99%	1 in 7,501	1 in 2,280,304
Andermann syndrome	SLC12A6	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Andermann syndrome	SLC12A6	AR	French Canadian	1 in 23	99%	1 in 2,201	1 in 202,492
Arginase deficiency	ARG1	AR	General	1 in 296	98%	1 in 14,751	<1 in 10 million
Argininosuccinate lyase deficiency	ASL	AR	General	1 in 132	90%	1 in 1,311	1 in 692,208
Aromatase deficiency	CYP19A1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Arthrogryposis, intellectual disability and seizures	SLC35A3	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Arthrogryposis, intellectual disability and seizures	SLC35A3	AR	Ashkenazi Jewish	1 in 453	98%	1 in 22,601	<1 in 10 million

AR autosomal recessive; XL X-linked.
*If patient not shown to be a carrier. ^ If patient not identified as a carrier and partner not tested for AR conditions

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Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
Arts syndrome	PRPS1	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
Asparagine synthetase deficiency	ASNS	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Aspartylglucosaminuria	AGA	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Aspartylglucosaminuria	AGA	AR	Finnish	1 in 71	98%	1 in 3,501	1 in 994,284
Ataxia neuropathy spectrum	POLG	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
Ataxia with isolated vitamin E deficiency	TTPA	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Ataxia with isolated vitamin E deficiency	TTPA	AR	Caucasian/European	1 in 267	90%	1 in 2,661	1 in 2,841,948
Ataxia-telangiectasia	ATM	AR	General	1 in 100	92%	1 in 1,239	1 in 495,600
Atelosteogenesis 2	SLC26A2	AR	General	1 in 158	90%	1 in 1,571	1 in 992,872
Atelosteogenesis 2	SLC26A2	AR	Finnish	1 in 50	90%	1 in 491	1 in 98,200
Autoimmune polyendocrinopathy syndrome, type 1	AIRE	AR	General	1 in 150	98%	1 in 7,451	1 in 4,470,600
Autoimmune polyendocrinopathy syndrome, type 1	AIRE		Finnish	1 in 79	98%	1 in 3,901	1 in 1,232,716
Autosomal recessive spastic ataxia of Charlevoix-Saguenay	SACS	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Autosomal recessive spastic ataxia of Charlevoix-Saguenay	SACS	AR	French Canadian	1 in 19	95%	1 in 361	1 in 27,436
Bardet-Biedl syndrome 11	TRIM32	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Bardet-Biedl syndrome 11	TRIM32	AR	Hutterite	1 in 12	98%	1 in 551	1 in 26,448
Bardet-Biedl syndrome 13	MKS1	AR	General	1 in 260	98%	1 in 12,951	<1 in 10 million
Bardet-Biedl syndrome 13	MKS1	AR	Finnish	1 in 47	98%	1 in 2,301	1 in 432,588
Bardet-Biedl syndrome 14	CEP290	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
Bardet-Biedl syndrome 2	BBS2	AR	General	1 in 621	99%	1 in 62,001	<1 in 10 million
Bardet-Biedl syndrome 2	BBS2	AR	Ashkenazi Jewish	1 in 107	99%	1 in 10,601	1 in 4,537,228
Bardet-Biedl syndrome type 1	BBS1	AR	General	1 in 367	99%	1 in 36,601	<1 in 10 million
Bardet-Biedl syndrome type 10	BBS10	AR	General	1 in 395	99%	1 in 39,401	<1 in 10 million
Bardet-Biedl syndrome type 12	BBS12	AR	General	1 in 791	99%	1 in 79,001	<1 in 10 million
Bare lymphocyte syndrome, type 2	CIITA	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Bartter syndrome	BSND	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Bernard-Soulier syndrome type A1	GP1BA	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Bernard-Soulier syndrome type C	GP9	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Beta thalassaemia	HBB	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
Beta thalassaemia	HBB	AR	African/African American	1 in 10	95%	1 in 181	1 in 7,240
Beta thalassaemia	HBB	AR	East Asian	1 in 50	95%	1 in 981	1 in 196,200
Beta thalassaemia	HBB	AR	Latino	1 in 128	95%	1 in 2,541	1 in 1,300,992
Beta thalassaemia	HBB	AR	Mediterranean	1 in 3	95%	1 in 41	1 in 492
Beta thalassaemia	HBB	AR	South Asian/Indian	1 in 25	95%	1 in 481	1 in 48,100
Bilateral frontoparietal polymicrogyria	ADGRG1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Biotinidase deficiency	BTD	AR	General	1 in 124	99%	1 in 12,301	1 in 6,101,296
Biotinidase deficiency	BTD	AR	Caucasian/European	1 in 71	99%	1 in 7,001	1 in 1,988,284
Biotinidase deficiency	BTD	AR	Latino	1 in 136	99%	1 in 13,501	1 in 7,344,544
Biotinidase deficiency	BTD	AR	Middle-Eastern	1 in 55	99%	1 in 5,401	1 in 1,188,220
Björnstad syndrome	BCS1L	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Bloom syndrome	BLM	AR	General	1 in 800	87%	1 in 6,147	<1 in 10 million
Bloom syndrome	BLM	AR	Ashkenazi Jewish	1 in 134	99%	1 in 13,301	1 in 7,129,336
Canavan disease	ASPA	AR	General	1 in 300	97%	1 in 9,968	<1 in 10 million
Canavan disease	ASPA	AR	Ashkenazi Jewish	1 in 55	96%	1 in 1,351	1 in 297,220
Carbamoyl phosphate synthetase 1 deficiency	CPS1	AR	General	1 in 570	98%	1 in 28,451	<1 in 10 million
Carnitine palmitoyltransferase 1A deficiency	CPT1A	AR	General	1 in 354	90%	1 in 3,531	1 in 4,999,896
Carnitine palmitoyltransferase 1A deficiency	CPT1A	AR	Hutterite	1 in 16	90%	1 in 151	1 in 9,664
Carnitine palmitoyltransferase 2 deficiency	CPT2	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Carnitine palmitoyltransferase 2 deficiency	CPT2	AR	Ashkenazi Jewish	1 in 51	95%	1 in 1,001	1 in 204,204
Carnitine-acylcarnitine translocase deficiency	SLC25A20	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Carpenter syndrome	RAB23	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Cartilage-hair hypoplasia	RMRP	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Cartilage-hair hypoplasia	RMRP	AR	Amish	1 in 16	99%	1 in 1,501	1 in 96,064
Cartilage-hair hypoplasia	RMRP	AR	Finnish	1 in 76	99%	1 in 7,501	1 in 2,280,304
Catecholaminergic polymorphic ventricular tachycardia, CASQ2-related	CASQ2	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
Catecholaminergic polymorphic ventricular tachycardia, TRDN-related	TRDN	AR	General	1 in 354	98%	1 in 17,651	<1 in 10 million
CEP290-related disorders	CEP290	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
Cerebrotendinous xanthomatosis	CYP27A1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Cerebrotendinous xanthomatosis	CYP27A1	AR	Moroccan Jewish	1 in 5	98%	1 in 201	1 in 4,020
Charcot-Marie-Tooth disease, GDAP1-related	GDAP1	AR	General	1 in 152	99%	1 in 15,101	1 in 9,181,408
Charcot-Marie-Tooth disease, SH3TC2-related	SH3TC2	AR	General	1 in 69	99%	1 in 6,801	1 in 1,877,076

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Charcot-Marie-Tooth disease, SURF1-related	SURF1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Charcot-Marie-Tooth disease, type 4B1	MTMR2	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Charcot-Marie-Tooth disease, type 4D	NDRG1	AR	General	1 in 22	98%	1 in 1,051	1 in 92,488
Charcot-Marie-Tooth disease, X-linked type 1	GJB1	XL	General	1 in 667	90%	1 in 6,661	1 in 26,644
Chediak-Higashi syndrome	LYST	AR	General	<1 in 500	90%	1 in 4,991	1 in 9,982,000
Childhood-onset severe retinal dystrophy, AIPL1-related	AIPL1	AR	General	1 in 409	99%	1 in 40,801	<1 in 10 million
Choreoacanthocytosis	VPS13A	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Choroideraemia	CHM	XL	General	1 in 25,000	95%	1 in 499,981	1 in 1,999,964
Chronic granulomatous disease	CYBA	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
Chronic granulomatous disease, X-linked	CYBB	XL	General	1 in 149,254	99%	1 in 14,925,301	<1 in 10 million
Citrin deficiency	SLC25A13	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Citrin deficiency	SLC25A13	AR	East Asian	1 in 65	95%	1 in 1,281	1 in 333,060
Citrullinaemia	ASS1	AR	General	1 in 119	96%	1 in 2,951	1 in 1,404,676
Citrullinaemia	ASS1	AR	East Asian	1 in 132	96%	1 in 3,276	1 in 1,729,728
COACH syndrome	RPGRIP1L	AR	General	1 in 259	98%	1 in 12,901	<1 in 10 million
Cockayne syndrome type A	ERCC8	AR	General	1 in 822	98%	1 in 41,051	<1 in 10 million
Cockayne syndrome type B	ERCC6	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
Cockayne syndrome type B	ERCC6	AR	Japanese	1 in 74	99%	1 in 7,301	1 in 2,161,096
Cohen syndrome	VPS13B	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Combined malonic and methylmalonic aciduria	ACSF3	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Combined oxidative phosphorylation deficiency, GFM1-related	GFM1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Combined oxidative phosphorylation deficiency, TSFM-related	TSFM	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Combined oxidative phosphorylation deficiency, TSFM-related	TSFM	AR	Finnish	1 in 80	98%	1 in 3,951	1 in 1,264,320
Combined pituitary hormone deficiency 2	PROP1	AR	General	1 in 45	98%	1 in 2,201	1 in 396,180
Combined pituitary hormone deficiency 3	LHX3	AR	General	1 in 45	98%	1 in 2,201	1 in 396,180
Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	CYP11B1	AR	General	1 in 158	98%	1 in 7,851	1 in 4,961,832
Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	CYP11B1	AR	Moroccan Jewish	1 in 35	98%	1 in 1,701	1 in 238,140
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	CYP17A1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	AR	General	1 in 61	99%	1 in 6,001	1 in 1,464,244
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	AR	Inuit	1 in 9	99%	1 in 801	1 in 28,836
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	AR	Middle-Eastern	1 in 35	99%	1 in 3,401	1 in 476,140
Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	HSD3B2	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Congenital adrenal hypoplasia, X-linked	NR0B1	XL	General	1 in 6,250	99%	1 in 624,901	1 in 2,499,804
Congenital amegakaryocytic thrombocytopaenia	MPL	AR	General	1 in 102	98%	1 in 5,051	1 in 2,060,808
Congenital amegakaryocytic thrombocytopaenia	MPL	AR	Ashkenazi Jewish	1 in 55	98%	1 in 2,701	1 in 594,220
Congenital disorder of glycosylation, type 1A	PMM2	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Congenital disorder of glycosylation, type 1A	PMM2	AR	Ashkenazi Jewish	1 in 57	99%	1 in 5,601	1 in 1,277,028
Congenital disorder of glycosylation, type 1A	PMM2	AR	Caucasian/European	1 in 71	99%	1 in 7,001	1 in 1,988,284
Congenital disorder of glycosylation, type 1B	MPI	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Congenital disorder of glycosylation, type 1C	ALG6	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Congenital hyperinsulinism	KCNJ11	AR	General	1 in 423	99%	1 in 42,201	<1 in 10 million
Congenital hyperinsulinism	KCNJ11	AR	Caucasian/European	1 in 232	99%	1 in 23,101	<1 in 10 million
Congenital hypothyroidism, DUOX2-related	DUOX2	AR	General	1 in 366	91%	1 in 4,057	1 in 5,938,797
Congenital hypothyroidism, DUOXA2-related	DUOXA2	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Congenital hypothyroidism, TSHB-related	TSHB	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
Congenital ichthyosis	TGM1	AR	General	1 in 224	95%	1 in 4,461	1 in 3,997,056
Congenital insensitivity to pain with anhidrosis	NTRK1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Congenital myasthenic syndrome, CHRNE-related	CHRNE	AR	General	1 in 408	99%	1 in 40,701	<1 in 10 million
Congenital myasthenic syndrome, RAPSN-related	RAPSN	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Congenital nephrotic syndrome, type 1	NPHS1	AR	General	1 in 289	98%	1 in 14,401	<1 in 10 million
Congenital nephrotic syndrome, type 1	NPHS1	AR	Finnish	1 in 50	98%	1 in 2,451	1 in 490,200
Congenital nephrotic syndrome, type 2	NPHS2	AR	General	1 in 289	98%	1 in 14,401	<1 in 10 million
Congenital nephrotic syndrome, type 2	NPHS2	AR	Finnish	1 in 50	98%	1 in 2,451	1 in 490,200

AR autosomal recessive; XL X-linked.
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Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
Congenital secretory chloride diarrhoea	SLC26A3	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Congenital secretory chloride diarrhoea	SLC26A3	AR	Middle-Eastern	1 in 57	98%	1 in 2,801	1 in 638,628
Corneal endothelial dystrophy	SLC4A11	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Corticosterone methyloxidase deficiency	CYP11B2	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Costeff syndrome	OPA3	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Costeff syndrome	OPA3	AR	Iraqi Jewish	1 in 50	98%	1 in 2,451	1 in 490,200
Creatine deficiency syndrome	SLC6A8	XL	General	1 in 3,434	98%	1 in 171,651	1 in 686,716
Crigler-Najjar syndrome	UGT1A1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Cystic fibrosis	CFTR	AR	General	1 in 32	99%	1 in 3,101	1 in 396,928
Cystic fibrosis	CFTR	AR	African/African American	1 in 61	99%	1 in 6,001	1 in 1,464,244
Cystic fibrosis	CFTR	AR	Ashkenazi Jewish	1 in 24	99%	1 in 2,301	1 in 220,896
Cystic fibrosis	CFTR	AR	Caucasian/European	1 in 25	99%	1 in 2,401	1 in 240,100
Cystic fibrosis	CFTR	AR	East Asian	1 in 94	99%	1 in 9,301	1 in 3,497,176
Cystic fibrosis	CFTR	AR	Latino	1 in 58	99%	1 in 5,701	1 in 1,322,632
Cystinosis	CTNS	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
Cystinosis	CTNS	AR	British	1 in 81	99%	1 in 8,001	1 in 2,592,324
D-bifunctional protein deficiency	HSD17B4	AR	General	1 in 158	98%	1 in 7,851	1 in 4,961,832
De Sanctis-Cacchione syndrome	ERCC6	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
De Sanctis-Cacchione syndrome	ERCC6	AR	Japanese	1 in 74	99%	1 in 7,301	1 in 2,161,096
Dent disease 2	OCRL	XL	General	1 in 250,000	95%	1 in 4,999,981	<1 in 10 million
Diastrophic dysplasia	SLC26A2	AR	General	1 in 158	90%	1 in 1,571	1 in 992,872
Diastrophic dysplasia	SLC26A2	AR	Finnish	1 in 50	90%	1 in 491	1 in 98,200
Dihydrolipoamide dehydrogenase deficiency	DLD	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Dihydrolipoamide dehydrogenase deficiency	DLD	AR	Ashkenazi Jewish	1 in 107	98%	1 in 5,301	1 in 2,268,828
Dihydropyrimidine dehydrogenase deficiency	DPYD	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Duchenne muscular dystrophy	DMD	XL	General	1 in 2,350	93%	1 in 33,558	1 in 134,260
Dyskeratosis congenita type 5	RTEL1	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
Dyskeratosis congenita type 5	RTEL1	AR	Ashkenazi Jewish	1 in 203	99%	1 in 20,201	<1 in 10 million
Dystrophic epidermolysis bullosa	COL7A1	AR	General	1 in 196	97%	1 in 6,501	1 in 5,096,784
Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	PLOD1	AR	General	1 in 159	99%	1 in 15,801	<1 in 10 million
Ehlers-Danlos syndrome, dermatosparaxis type 7C	ADAMTS2	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Ehlers-Danlos syndrome, dermatosparaxis type 7C	ADAMTS2	AR	Ashkenazi Jewish	1 in 248	98%	1 in 12,351	<1 in 10 million
Ellis-van Creveld syndrome, EVC2-related	EVC2	AR	General	1 in 240	98%	1 in 11,951	<1 in 10 million
Ellis-van Creveld syndrome, EVC2-related	EVC2	AR	Amish	1 in 7	98%	1 in 301	1 in 8,428
Ellis-van Creveld syndrome, EVC-related	EVC	AR	General	1 in 142	98%	1 in 7,051	1 in 4,004,968
Ellis-van Creveld syndrome, EVC-related	EVC	AR	Amish	1 in 7	98%	1 in 301	1 in 8,428
Emery-Dreifuss muscular dystrophy	EMD	XL	General	1 in 81,967	99%	1 in 8,196,601	<1 in 10 million
Enhanced S-cone syndrome	NR2E3	AR	General	1 in 209	98%	1 in 10,401	1 in 8,695,236
Ethylmalonic encephalopathy	ETHE1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Fabry disease	GLA	XL	General	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
Factor 11 deficiency	F11	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Factor 11 deficiency	F11	AR	Ashkenazi Jewish	1 in 11	98%	1 in 501	1 in 22,044
Familial dysautonomia	ELP1	AR	General	1 in 300	99%	1 in 29,901	<1 in 10 million
Familial dysautonomia	ELP1	AR	Ashkenazi Jewish	1 in 31	99%	1 in 3,001	1 in 372,124
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	General	1 in 112	98%	1 in 5,551	1 in 2,486,848
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	Ashkenazi Jewish	1 in 44	98%	1 in 2,151	1 in 378,576
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	Finnish	1 in 25	98%	1 in 1,201	1 in 120,100
Familial hyperinsulinism, ABCC8-related	ABCC8	AR	Middle-Eastern	1 in 25	98%	1 in 1,201	1 in 120,100
Familial lipoprotein lipase deficiency	LPL	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
Familial lipoprotein lipase deficiency	LPL	AR	French Canadian	1 in 46	99%	1 in 4,501	1 in 828,184
Fanconi anaemia group A	FANCA	AR	General	1 in 239	98%	1 in 11,901	<1 in 10 million
Fanconi anaemia group C	FANCC	AR	General	1 in 535	99%	1 in 53,401	<1 in 10 million
Fanconi anaemia group C	FANCC	AR	Ashkenazi Jewish	1 in 99	99%	1 in 9,801	1 in 3,881,196
Fanconi anaemia group G	FANCG	AR	General	1 in 632	90%	1 in 6,311	<1 in 10 million
Fetal akinesia deformation sequence	RAPSN	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Fragile X syndrome	FMR1	XL	General	1 in 151	99%	1 in 15,001	1 in 60,004
Fragile X syndrome	FMR1	XL	Ashkenazi Jewish	1 in 115	99%	1 in 11,401	1 in 45,604
Fukuyama congenital muscular dystrophy	FKTN	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Fukuyama congenital muscular dystrophy	FKTN	AR	Ashkenazi Jewish	1 in 150	99%	1 in 14,901	1 in 8,940,600
Fukuyama congenital muscular dystrophy	FKTN	AR	Japanese	1 in 82	99%	1 in 8,101	1 in 2,657,128
Fumarase deficiency	FH	AR	General	<1 in 500	90%	1 in 4,991	1 in 9,982,000
Galactokinase deficiency	GALK1	AR	General	1 in 110	95%	1 in 2,181	1 in 959,640
Galactokinase deficiency	GALK1	AR	Irish	1 in 64	95%	1 in 1,261	1 in 322,816
Galactosaemia	GALT	AR	General	1 in 110	95%	1 in 2,181	1 in 959,640

AR autosomal recessive; XL X-linked.
*If patient not shown to be a carrier. ^ If patient not identified as a carrier and partner not tested for AR conditions

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Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
Galactosaemia	GALT	AR	African/African American	1 in 94	95%	1 in 1,861	1 in 699,736
Galactose epimerase deficiency	GALE	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Gaucher disease	GBA	AR	General	1 in 77	99%	1 in 7,601	1 in 2,341,108
Gaucher disease	GBA	AR	African/African American	1 in 35	99%	1 in 3,401	1 in 476,140
Gaucher disease	GBA	AR	Ashkenazi Jewish	1 in 15	99%	1 in 1,401	1 in 84,060
Gitelman syndrome	SLC12A3	AR	General	1 in 100	98%	1 in 4,951	1 in 1,980,400
Glutamate formiminotransferase deficiency	FTCD	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Glutaric aciduria, type 1	GCDH	AR	General	1 in 87	98%	1 in 4,301	1 in 1,496,748
Glutaric aciduria, type 1	GCDH	AR	Amish	1 in 9	98%	1 in 401	1 in 14,436
Glutaric aciduria, type 2A	ETFA	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Glutaric aciduria, type 2B	ETFB	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Glutaric aciduria, type 2C	ETFDH	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
Glutaric aciduria, type 2C	ETFDH	AR	East Asian	1 in 74	98%	1 in 3,651	1 in 1,080,696
Glycine encephalopathy, AMT-related	AMT	AR	General	1 in 373	98%	1 in 18,601	<1 in 10 million
Glycine encephalopathy, AMT-related	AMT	AR	Finnish	1 in 117	98%	1 in 5,801	1 in 2,714,868
Glycine encephalopathy, GLDC-related	GLDC	AR	General	1 in 193	98%	1 in 9,601	1 in 7,411,972
Glycine encephalopathy, GLDC-related	GLDC	AR	British Columbia Canadian	1 in 125	99%	1 in 12,401	1 in 6,200,500
Glycine encephalopathy, GLDC-related	GLDC	AR	Finnish	1 in 117	99%	1 in 11,601	1 in 5,429,268
Glycogen storage disease, type 1A	G6PC	AR	General	1 in 177	95%	1 in 3,521	1 in 2,492,868
Glycogen storage disease, type 1A	G6PC	AR	Ashkenazi Jewish	1 in 64	95%	1 in 1,261	1 in 322,816
Glycogen storage disease, type 1B	SLC37A4	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
Glycogen storage disease, type 1B	SLC37A4	AR	Ashkenazi Jewish	1 in 71	95%	1 in 1,401	1 in 397,884
Glycogen storage disease, type 3	AGL	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
Glycogen storage disease, type 3	AGL	AR	Faroese	1 in 28	95%	1 in 541	1 in 60,592
Glycogen storage disease, type 3	AGL	AR	Inuit	1 in 25	95%	1 in 481	1 in 48,100
Glycogen storage disease, type 3	AGL	AR	North African Jewish	1 in 37	95%	1 in 721	1 in 106,708
Glycogen storage disease, type 4	GBE1	AR	General	1 in 387	99%	1 in 38,601	<1 in 10 million
Glycogen storage disease, type 5	PYGM	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Glycogen storage disease, type 5	PYGM	AR	Caucasian/European	1 in 206	99%	1 in 20,501	<1 in 10 million
Glycogen storage disease, type 7	PFKM	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
GM1-gangliosidosis	GLB1	AR	General	1 in 134	99%	1 in 13,301	1 in 7,129,336
GM1-gangliosidosis	GLB1	AR	Maltese	1 in 30	99%	1 in 2,901	1 in 348,120
GM1-gangliosidosis	GLB1	AR	Roma	1 in 50	99%	1 in 4,901	1 in 980,200
GRACILE syndrome	BCS1L	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Guanidinoacetate methyltransferase deficiency	GAMT	AR	General	1 in 371	99%	1 in 37,001	<1 in 10 million
Gyrate atrophy of choroid and retina	OAT	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Haemochromatosis, type 2A	HJV	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
Haemochromatosis, type 3	TFR2	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Haemophilia A	F8	XL	General	1 in 3,250	48%	1 in 6,249	1 in 25,000
Haemophilia B	F9	XL	General	1 in 15,000	99%	1 in 1,499,901	1 in 5,999,804
Hartnup disorder	SLC6A19	AR	General	1 in 87	99%	1 in 8,601	1 in 2,993,148
Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	MPV17	AR	General	<1 in 500	96%	1 in 12,476	<1 in 10 million
Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	MPV17	AR	Native American	1 in 20	96%	1 in 476	1 in 38,080
Hereditary folate malabsorption	SLC46A1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Hereditary folate malabsorption	SLC46A1	AR	Puerto Rican	1 in 500	99%	1 in 49,901	<1 in 10 million
Hereditary fructose intolerance	ALDOB	AR	General	1 in 122	99%	1 in 12,101	1 in 5,905,288
Hereditary fructose intolerance	ALDOB	AR	African/African American	1 in 250	99%	1 in 24,901	<1 in 10 million
Hereditary fructose intolerance	ALDOB	AR	Caucasian/European	1 in 67	99%	1 in 6,601	1 in 1,769,068
Hereditary fructose intolerance	ALDOB	AR	Middle-Eastern	1 in 97	99%	1 in 9,601	1 in 3,725,188
Heřmanský-Pudlák syndrome 1	HPS1	AR	General	1 in 354	98%	1 in 17,651	<1 in 10 million
Heřmanský-Pudlák syndrome 1	HPS1	AR	Puerto Rican	1 in 21	98%	1 in 1,001	1 in 84,084
Heřmanský-Pudlák syndrome 3	HPS3	AR	General	1 in 354	98%	1 in 17,651	<1 in 10 million
Holocarboxylase synthetase deficiency	HLCS	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Homocystinuria due to cystathionine beta-synthase deficiency	CBS	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
Homocystinuria due to cystathionine beta-synthase deficiency	CBS	AR	Caucasian/European	1 in 86	99%	1 in 8,501	1 in 2,924,344
Homocystinuria due to cystathionine beta-synthase deficiency	CBS	AR	Middle-Eastern	1 in 21	99%	1 in 2,001	1 in 168,084
Homocystinuria-megaloblastic anaemia, cobalamin E type	MTRR	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Hydrolethalus syndrome	HYLS1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Hydrolethalus syndrome	HYLS1	AR	Finnish	1 in 50	98%	1 in 2,451	1 in 490,200

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Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
Hyper IgM syndrome, X-linked	CD40LG	XL	General	1 in 50,000	98%	1 in 2,499,951	1 in 9,999,904
Hyperimmunoglobulinaemia D syndrome	MVK	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Hypermethioninaemia due to adenosine kinase deficiency	ADK	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Hypermethioninaemia due to deficiency of S-adenosylhomocysteine hydrolase	AHCY	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Hyperornithinaemia hyperammonaemia homocitrullinuria syndrome (Triple H syndrome)	SLC25A15	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Hyperornithinaemia hyperammonaemia homocitrullinuria syndrome (Triple H syndrome)	SLC25A15	AR	French Canadian	1 in 37	99%	1 in 3,601	1 in 532,948
Hyperprolinaemia, type 2	ALDH4A1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Hypogonadotropic hypogonadism, GNRHR-related	GNRHR	AR	General	1 in 347	99%	1 in 34,601	<1 in 10 million
Hypohidrotic ectodermal dysplasia	EDA	XL	General	1 in 14,167	99%	1 in 1,416,601	1 in 5,666,472
Hypomyelinating leucodystrophy, POLR1C-related	POLR1C	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Hypophosphatasia	ALPL	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
Hypophosphatasia	ALPL	AR	Caucasian/European	1 in 274	95%	1 in 5,461	1 in 5,985,256
Hypophosphatasia	ALPL	AR	Mennonite	1 in 25	95%	1 in 481	1 in 48,100
Inclusion body myopathy type 2 (Nonaka myopathy)	GNE	AR	General	<1 in 500	80%	1 in 2,496	1 in 4,992,000
Inclusion body myopathy type 2 (Nonaka myopathy)	GNE	AR	Iranian Jewish	1 in 11	80%	1 in 51	1 in 2,244
Infantile neuroaxonal dystrophy	PLA2G6	AR	General	1 in 500	97%	1 in 16,634	<1 in 10 million
Isovaleric acidaemia	IVD	AR	General	1 in 167	90%	1 in 1,661	1 in 1,109,548
Isovaleric acidaemia	IVD	AR	African/African American	1 in 100	90%	1 in 991	1 in 396,400
Isovaleric acidaemia	IVD	AR	Caucasian/European	1 in 115	90%	1 in 1,141	1 in 524,860
Isovaleric acidaemia	IVD	AR	East Asian	1 in 407	90%	1 in 4,061	1 in 6,611,308
Joubert syndrome 2	TMEM216	AR	General	1 in 141	98%	1 in 7,001	1 in 3,948,564
Joubert syndrome 2	TMEM216	AR	Ashkenazi Jewish	1 in 92	98%	1 in 4,551	1 in 1,674,768
Joubert syndrome 28	MKS1	AR	General	1 in 260	98%	1 in 12,951	<1 in 10 million
Joubert syndrome 28	MKS1	AR	Finnish	1 in 47	98%	1 in 2,301	1 in 432,588
Joubert syndrome 4	NPHP1	AR	General	1 in 480	98%	1 in 23,951	<1 in 10 million
Joubert syndrome 4	NPHP1	AR	Finnish	1 in 124	98%	1 in 6,151	1 in 3,050,896
Joubert syndrome 5	CEP290	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
Joubert syndrome 7	RPGRIP1L	AR	General	1 in 259	98%	1 in 12,901	<1 in 10 million
Joubert syndrome, AHI1-related	AHI1	AR	General	1 in 448	99%	1 in 44,701	<1 in 10 million
Joubert syndrome, ARL13B-related	ARL13B	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Junctional epidermolysis bullosa, LAMA3-related	LAMA3	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
Junctional epidermolysis bullosa, LAMB3-related	LAMB3	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
Junctional epidermolysis bullosa, LAMC2-related	LAMC2	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
Juvenile retinoschisis, X-linked	RS1	XL	General	1 in 2,500	96%	1 in 62,476	1 in 249,956
Krabbe disease	GALC	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
Krabbe disease	GALC	AR	Israeli Druze	1 in 6	99%	1 in 501	1 in 12,024
L1 syndrome	L1CAM	XL	General	1 in 15,000	99%	1 in 1,499,901	1 in 5,999,804
Laryngo-onycho-cutaneous syndrome	LAMA3	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
Leber congenital amaurosis 10	CEP290	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
Leber congenital amaurosis 2	RPE65	AR	General	1 in 228	98%	1 in 11,351	<1 in 10 million
Leber congenital amaurosis 5	LCA5	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Leber congenital amaurosis 8	CRB1	AR	General	1 in 104	98%	1 in 5,151	1 in 2,142,816
Leber congenital amaurosis, type 13	RDH12	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Leber congenital amaurosis, type 13	RDH12	AR	Caucasian/European	1 in 456	98%	1 in 22,751	<1 in 10 million
Leigh syndrome with complex 4 deficiency	LRPPRC	AR	General	1 in 447	98%	1 in 22,301	<1 in 10 million
Leigh syndrome with complex 4 deficiency	LRPPRC	AR	Faroese	1 in 21	98%	1 in 1,001	1 in 84,084
Leigh syndrome with complex 4 deficiency	LRPPRC	AR	French Canadian	1 in 22	98%	1 in 1,051	1 in 92,488
Leigh syndrome, SURF1-related	SURF1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Lethal congenital contracture syndrome 1	GLE1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Lethal congenital contracture syndrome 1	GLE1	AR	Finnish	1 in 80	98%	1 in 3,951	1 in 1,264,320
Leucoencephalopathy with vanishing white matter	EIF2B5	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Limb-girdle muscular dystrophy, type 2A	CAPN3	AR	Caucasian/European	1 in 103	98%	1 in 5,101	1 in 2,101,612
Limb-girdle muscular dystrophy, type 2B	DYSF	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Limb-girdle muscular dystrophy, type 2B	DYSF	AR	Japanese	1 in 332	95%	1 in 6,621	1 in 8,792,688
Limb-girdle muscular dystrophy, type 2B	DYSF	AR	Libyan Jewish	1 in 18	95%	1 in 341	1 in 24,552
Limb-girdle muscular dystrophy, type 2C	SGCG	AR	General	1 in 381	98%	1 in 19,001	<1 in 10 million
Limb-girdle muscular dystrophy, type 2C	SGCG	AR	Moroccan	1 in 250	98%	1 in 12,451	<1 in 10 million
Limb-girdle muscular dystrophy, type 2C	SGCG	AR	Roma/Gypsy	1 in 96	98%	1 in 4,751	1 in 1,824,384
Limb-girdle muscular dystrophy, type 2D	SGCA	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Limb-girdle muscular dystrophy, type 2D	SGCA	AR	Caucasian/European	1 in 288	98%	1 in 14,351	<1 in 10 million
Limb-girdle muscular dystrophy, type 2D	SGCA	AR	Finnish	1 in 150	98%	1 in 7,451	1 in 4,470,600

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Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
Limb-girdle muscular dystrophy, type 2E	SGCB	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Limb-girdle muscular dystrophy, type 2E	SGCB	AR	Caucasian/European	1 in 406	98%	1 in 20,251	<1 in 10 million
Limb-girdle muscular dystrophy, type 2F	SGCD	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Limb-girdle muscular dystrophy, type 2H	TRIM32	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Limb-girdle muscular dystrophy, type 2H	TRIM32	AR	Hutterite	1 in 12	98%	1 in 551	1 in 26,448
Lipoid congenital adrenal hyperplasia	STAR	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Lissencephaly, X-linked	DCX	XL	General	1 in 42,500	98%	1 in 2,124,951	1 in 8,499,904
Liver failure, acute infantile	TRMU	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Liver failure, acute infantile	TRMU	AR	Yemeni Jewish	1 in 34	98%	1 in 1,651	1 in 224,536
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	HADHA	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	HADHA	AR	Finnish	1 in 124	98%	1 in 6,151	1 in 3,050,896
Lowe syndrome	OCRL	XL	General	1 in 250,000	95%	1 in 4,999,981	<1 in 10 million
Lujan-Fryns syndrome, UPF3B-related	UPF3B	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
Lujan-Fryns syndrome, ZDHHC9-related	ZDHHC9	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
Lysinuric protein intolerance	SLC7A7	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Lysinuric protein intolerance	SLC7A7	AR	Finnish	1 in 122	95%	1 in 2,421	1 in 1,181,448
Lysinuric protein intolerance	SLC7A7	AR	Japanese	1 in 119	95%	1 in 2,361	1 in 1,123,836
Lysosomal acid lipase deficiency	LIPA	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Lysosomal acid lipase deficiency	LIPA	AR	Caucasian/European	1 in 112	99%	1 in 11,101	1 in 4,973,248
Macular corneal dystrophy, CHST6-related	CHST6	AR	General	1 in 79	99%	1 in 7,801	1 in 2,465,116
Maple syrup urine disease, type 1A	BCKDHA	AR	General	1 in 321	98%	1 in 16,001	<1 in 10 million
Maple syrup urine disease, type 1A	BCKDHA	AR	Mennonite	1 in 10	98%	1 in 451	1 in 18,040
Maple syrup urine disease, type 1B	BCKDHB	AR	General	1 in 364	98%	1 in 18,151	<1 in 10 million
Maple syrup urine disease, type 1B	BCKDHB	AR	Ashkenazi Jewish	1 in 97	98%	1 in 4,801	1 in 1,862,788
Maple syrup urine disease, type 2	DBT	AR	General	1 in 481	98%	1 in 24,001	<1 in 10 million
Meckel syndrome 1	MKS1	AR	General	1 in 260	98%	1 in 12,951	<1 in 10 million
Meckel syndrome 1	MKS1	AR	Finnish	1 in 47	98%	1 in 2,301	1 in 432,588
Meckel syndrome 2	TMEM216	AR	General	1 in 141	98%	1 in 7,001	1 in 3,948,564
Meckel syndrome 2	TMEM216	AR	Ashkenazi Jewish	1 in 92	98%	1 in 4,551	1 in 1,674,768
Meckel syndrome 4	CEP290	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
Meckel syndrome 5	RPGRIP1L	AR	General	1 in 259	98%	1 in 12,901	<1 in 10 million
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM	AR	General	1 in 69	98%	1 in 3,401	1 in 938,676
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM	AR	Caucasian/European	1 in 52	99%	1 in 5,101	1 in 1,061,008
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM	AR	East Asian	1 in 198	99%	1 in 19,701	<1 in 10 million
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM	AR	Native American	1 in 43	96%	1 in 1,051	1 in 180,772
Megalencephalic leucoencephalopathy with subcortical cysts	MLC1	AR	General	<1 in 500	97%	1 in 16,634	<1 in 10 million
Menkes disease	ATP7A	XL	General	1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
Metachromatic leucodystrophy	ARSA	AR	General	1 in 100	95%	1 in 1,981	1 in 792,400
Metachromatic leucodystrophy	ARSA	AR	Caucasian/European	1 in 78	95%	1 in 1,541	1 in 480,792
Metachromatic leucodystrophy due to saposin B deficiency	PSAP	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Metaphyseal dysplasia without hypotrichosis	RMRP	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Metaphyseal dysplasia without hypotrichosis	RMRP	AR	Amish	1 in 16	99%	1 in 1,501	1 in 96,064
Metaphyseal dysplasia without hypotrichosis	RMRP	AR	Finnish	1 in 76	99%	1 in 7,501	1 in 2,280,304
Methylmalonic acidaemia, MUT-related	MUT	AR	General	1 in 195	96%	1 in 4,851	1 in 3,783,780
Methylmalonic acidaemia, MUT-related	MUT	AR	East Asian	1 in 53	96%	1 in 1,301	1 in 275,812
Methylmalonic acidaemia, MUT-related	MUT	AR	Middle-Eastern	1 in 52	96%	1 in 1,276	1 in 265,408
Methylmalonic aciduria and homocystinuria, cblC type	MMACHC	AR	General	1 in 134	90%	1 in 1,331	1 in 713,416
Methylmalonic aciduria and homocystinuria, cblD type	MMADHC	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Methylmalonic aciduria and homocystinuria, cblF type	LMBRD1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Methylmalonic aciduria and homocystinuria, cblJ type	ABCD4	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Methylmalonic aciduria, cblA type	MMAA	AR	General	1 in 301	97%	1 in 10,001	<1 in 10 million
Methylmalonic aciduria, cblB type	MMAB	AR	General	1 in 435	98%	1 in 21,701	<1 in 10 million
Methylmalonyl-CoA epimerase deficiency	MCEE	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Mevalonate kinase deficiency	MVK	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Microphthalmia with or without coloboma	VSX2	AR	General	1 in 91	98%	1 in 4,501	1 in 1,638,364
Microphthalmia, isolated 3	RAX	AR	General	1 in 289	99%	1 in 28,801	<1 in 10 million

AR autosomal recessive; XL X-linked.
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Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
Mitochondrial complex 1 deficiency (Leigh syndrome), NDUFAF5-related	NDUFAF5	AR	General	1 in 447	98%	1 in 22,301	<1 in 10 million
Mitochondrial complex 1 deficiency (Leigh syndrome), NDUFAF5-related	NDUFAF5	AR	Ashkenazi Jewish	1 in 290	98%	1 in 14,451	<1 in 10 million
Mitochondrial complex 1 deficiency (Leigh syndrome), NDUFS6-related	NDUFS6	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Mitochondrial complex 3 deficiency	BCS1L	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Mitochondrial myopathy and sideroblastic anaemia 1	PUS1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	TYMP	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Mucolipidosis 2 alpha/beta	GNPTAB	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Mucolipidosis 3 alpha/beta	GNPTAB	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Mucolipidosis 3 gamma	GNPTG	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Mucolipidosis 4	MCOLN1	AR	General	1 in 300	99%	1 in 29,901	<1 in 10 million
Mucolipidosis 4	MCOLN1	AR	Ashkenazi Jewish	1 in 100	99%	1 in 9,901	1 in 3,960,400
Mucopolysaccharidosis, type 1 (Hurler syndrome)	IDUA	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Mucopolysaccharidosis, type 1 (Hurler syndrome)	IDUA	AR	Caucasian/European	1 in 153	95%	1 in 3,041	1 in 1,861,092
Mucopolysaccharidosis, type 2 (Hunter syndrome)	IDS	XL	General	1 in 50,000	91%	1 in 555,545	1 in 2,222,204
Mucopolysaccharidosis, type 3A (Sanfilippo syndrome A)	SGSH	AR	General	1 in 454	98%	1 in 22,651	<1 in 10 million
Mucopolysaccharidosis, type 3A (Sanfilippo syndrome A)	SGSH	AR	Caucasian/European	1 in 253	98%	1 in 12,601	<1 in 10 million
Mucopolysaccharidosis, type 3B (Sanfilippo syndrome B)	NAGLU	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Mucopolysaccharidosis, type 3B (Sanfilippo syndrome B)	NAGLU	AR	Caucasian/European	1 in 346	99%	1 in 34,501	<1 in 10 million
Mucopolysaccharidosis, type 3B (Sanfilippo syndrome B)	NAGLU	AR	East Asian	1 in 298	99%	1 in 29,701	<1 in 10 million
Mucopolysaccharidosis, type 3C (Sanfilippo syndrome C)	HGSNAT	AR	General	1 in 434	98%	1 in 21,651	<1 in 10 million
Mucopolysaccharidosis, type 3C (Sanfilippo syndrome C)	HGSNAT	AR	Caucasian/European	1 in 345	98%	1 in 17,201	<1 in 10 million
Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	GNS	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Mucopolysaccharidosis, type 4A (Morquio syndrome A)	GALNS	AR	General	1 in 224	97%	1 in 7,434	1 in 6,660,864
Mucopolysaccharidosis, type 4B (Morquio syndrome B)	GLB1	AR	General	1 in 134	99%	1 in 13,301	1 in 7,129,336
Mucopolysaccharidosis, type 4B (Morquio syndrome B)	GLB1	AR	Maltese	1 in 30	99%	1 in 2,901	1 in 348,120
Mucopolysaccharidosis, type 4B (Morquio syndrome B)	GLB1	AR	Roma	1 in 50	99%	1 in 4,901	1 in 980,200
Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	ARSB	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	ARSB	AR	Western Australian	1 in 283	98%	1 in 14,101	<1 in 10 million
Mucopolysaccharidosis, type 7	GUSB	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
Mucopolysaccharidosis, type 9	HYAL1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Multiple epiphyseal dysplasia	SLC26A2	AR	General	1 in 158	90%	1 in 1,571	1 in 992,872
Multiple epiphyseal dysplasia	SLC26A2	AR	Finnish	1 in 50	90%	1 in 491	1 in 98,200
Multiple pterygium syndrome	CHRNG	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Multiple sulphatase deficiency	SUMF1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Multiple sulphatase deficiency	SUMF1	AR	Ashkenazi Jewish	1 in 320	98%	1 in 15,951	<1 in 10 million
Muscular dystrophy, LAMA2-related	LAMA2	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Muscular dystrophy, LAMA2-related	LAMA2	AR	Caucasian/European	1 in 125	99%	1 in 12,401	1 in 6,200,500
Muscular dystrophy-dystroglycanopathy	POMGNT1	AR	General	1 in 462	98%	1 in 23,051	<1 in 10 million
Muscular dystrophy-dystroglycanopathy	POMGNT1	AR	Finnish	1 in 111	98%	1 in 5,501	1 in 2,442,444
Muscular dystrophy-dystroglycanopathy, FKRP-related	FKRP	AR	General	1 in 158	98%	1 in 7,851	1 in 4,961,832
Muscular dystrophy-dystroglycanopathy, FKTN-related	FKTN	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Muscular dystrophy-dystroglycanopathy, FKTN-related	FKTN	AR	Ashkenazi Jewish	1 in 150	99%	1 in 14,901	1 in 8,940,600
Muscular dystrophy-dystroglycanopathy, FKTN-related	FKTN	AR	Japanese	1 in 82	99%	1 in 8,101	1 in 2,657,128
Muscular dystrophy-dystroglycanopathy, POMT1-related	POMT1	AR	General	1 in 290	99%	1 in 28,901	<1 in 10 million
Muscular dystrophy-dystroglycanopathy, POMT2-related	POMT2	AR	General	1 in 371	99%	1 in 37,001	<1 in 10 million
Myocerebrohepatopathy syndrome	POLG	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
Myotubular myopathy, X-linked	MTM1	XL	General	1 in 25,000	98%	1 in 1,249,951	1 in 4,999,904
N-acetylglutamate synthase deficiency	NAGS	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Nemaline myopathy	NEB	AR	General	1 in 112	98%	1 in 5,551	1 in 2,486,848
Nemaline myopathy	NEB	AR	Amish	1 in 11	98%	1 in 501	1 in 22,044
Nemaline myopathy	NEB	AR	Ashkenazi Jewish	1 in 108	98%	1 in 5,351	1 in 2,311,632
Nemaline myopathy	NEB	AR	Finnish	1 in 112	98%	1 in 5,551	1 in 2,486,848
Nephrogenic diabetes insipidus	AQP2	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Nephrogenic diabetes insipidus	AQP2	AR	Finnish	1 in 169	95%	1 in 3,361	1 in 2,272,036
Nephronophthisis	NPHP1	AR	General	1 in 480	98%	1 in 23,951	<1 in 10 million
Nephronophthisis	NPHP1	AR	Finnish	1 in 124	98%	1 in 6,151	1 in 3,050,896
Neuronal ceroid lipofuscinosis, CLN3-related	CLN3	AR	General	1 in 230	98%	1 in 11,451	<1 in 10 million
Neuronal ceroid lipofuscinosis, CLN3-related	CLN3	AR	Finnish	1 in 72	98%	1 in 3,551	1 in 1,022,688
Neuronal ceroid lipofuscinosis, CLN5-related	CLN5	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Neuronal ceroid lipofuscinosis, CLN5-related	CLN5	AR	Finnish	1 in 115	95%	1 in 2,281	1 in 1,049,260
Neuronal ceroid lipofuscinosis, CLN6-related	CLN6	AR	General	<1 in 500	92%	1 in 6,239	<1 in 10 million
Neuronal ceroid lipofuscinosis, CLN8-related	CLN8	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million

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Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
Neuronal ceroid lipofuscinosis, CLN8-related	CLN8	AR	Finnish	1 in 135	95%	1 in 2,681	1 in 1,447,740
Neuronal ceroid lipofuscinosis, MFSD8-related	MFSD8	AR	General	<1 in 500	95%	1 in 9,981	<1 in 10 million
Neuronal ceroid lipofuscinosis, PPT1-related	PPT1	AR	General	1 in 368	98%	1 in 18,351	<1 in 10 million
Neuronal ceroid lipofuscinosis, PPT1-related	PPT1	AR	Caucasian/European	1 in 488	98%	1 in 24,351	<1 in 10 million
Neuronal ceroid lipofuscinosis, PPT1-related	PPT1	AR	Finnish	1 in 75	98%	1 in 3,701	1 in 1,110,300
Neuronal ceroid lipofuscinosis, TPP1-related	TPP1	AR	General	1 in 252	97%	1 in 8,368	1 in 8,434,944
Neuronal ceroid lipofuscinosis, TPP1-related	TPP1	AR	French Canadian	1 in 53	97%	1 in 1,734	1 in 367,608
Niemann-Pick disease, type A/B	SMPD1	AR	General	1 in 250	95%	1 in 4,981	1 in 4,981,000
Niemann-Pick disease, type A/B	SMPD1	AR	Ashkenazi Jewish	1 in 115	95%	1 in 2,281	1 in 1,049,260
Niemann-Pick disease, type A/B	SMPD1	AR	Latino	1 in 106	95%	1 in 2,101	1 in 890,824
Niemann-Pick disease, type C1	NPC1	AR	General	1 in 194	90%	1 in 1,931	1 in 1,498,456
Niemann-Pick disease, type C2	NPC2	AR	General	1 in 194	99%	1 in 19,301	<1 in 10 million
Nijmegen breakage syndrome	NBN	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
Non-syndromic hearing loss, GJB2-related	GJB2	AR	General	1 in 42	99%	1 in 4,101	1 in 688,968
Non-syndromic hearing loss, GJB2-related	GJB2	AR	African/African American	1 in 25	99%	1 in 2,401	1 in 240,100
Non-syndromic hearing loss, GJB2-related	GJB2	AR	Ashkenazi Jewish	1 in 21	99%	1 in 2,001	1 in 168,084
Non-syndromic hearing loss, GJB2-related	GJB2	AR	Caucasian/European	1 in 33	99%	1 in 3,201	1 in 422,532
Non-syndromic hearing loss, GJB2-related	GJB2	AR	Latino	1 in 100	99%	1 in 9,901	1 in 3,960,400
Non-syndromic hearing loss, GJB2-related	GJB2	AR	Middle-Eastern	1 in 83	99%	1 in 8,201	1 in 2,722,732
Non-syndromic hearing loss, GJB2-related	GJB2	AR	South Asian/Indian	1 in 148	99%	1 in 14,701	1 in 8,702,992
Non-syndromic hearing loss, GJB6-related	GJB6	AR	General	1 in 423	99%	1 in 42,201	<1 in 10 million
Non-syndromic hearing loss, LOXHD1-related	LOXHD1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Non-syndromic hearing loss, LOXHD1-related	LOXHD1	AR	Ashkenazi Jewish	1 in 180	98%	1 in 8,951	1 in 6,444,720
Non-syndromic hearing loss, MYO7A-related	MYO7A	AR	General	1 in 206	98%	1 in 10,251	1 in 8,446,824
Non-syndromic hearing loss, MYO7A-related	MYO7A	AR	East Asian	1 in 62	98%	1 in 3,051	1 in 756,648
Non-syndromic hearing loss, OTOF-related	OTOF	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Non-syndromic hearing loss, OTOF-related	OTOF	AR	Spanish	1 in 106	99%	1 in 10,501	1 in 4,452,424
Non-syndromic hearing loss, PCDH15-related	PCDH15	AR	General	1 in 395	98%	1 in 19,701	1 in 78,804
Non-syndromic hearing loss, PCDH15-related	PCDH15	AR	Ashkenazi Jewish	1 in 72	98%	1 in 3,551	1 in 14,204
Non-syndromic hearing loss, PRPS1-related	PRPS1	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
Non-syndromic hearing loss, USH1C-related	USH1C	AR	General	1 in 353	90%	1 in 3,521	1 in 4,971,652
Non-syndromic hearing loss, USH1C-related	USH1C	AR	French Canadian	1 in 227	90%	1 in 2,261	1 in 2,052,988
Norrie disease	NDP	XL	General	<1 in 50,000	98%	1 in 2,499,951	<1 in 10 million
Odontoonychodermal dysplasia	WNT10A	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Omenn syndrome, RAG1-related	RAG1	AR	General	1 in 137	98%	1 in 6,801	1 in 3,726,948
Omenn syndrome, RAG2-related	RAG2	AR	General	1 in 137	98%	1 in 6,801	1 in 3,726,948
Ornithine transcarbamylase deficiency	OTC	XL	General	1 in 7,000	90%	1 in 69,991	1 in 279,984
Osteogenesis imperfecta, type 8	P3H1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Osteogenesis imperfecta, type 8	P3H1	AR	West African	1 in 67	99%	1 in 6,601	1 in 1,769,068
Osteogenesis imperfecta, type 8	P3H1	AR	African American	1 in 250	99%	1 in 24,901	<1 in 10,000,000
Osteopetrosis, TCIRG1-related	TCIRG1	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
Pantothenate kinase-associated neurodegeneration	PANK2	AR	General	1 in 289	99%	1 in 28,801	<1 in 10 million
Pendred syndrome	SLC26A4	AR	General	1 in 80	98%	1 in 3,951	1 in 1,264,320
Pendred syndrome	SLC26A4	AR	African/African American	1 in 76	98%	1 in 3,751	1 in 1,140,304
Pendred syndrome	SLC26A4	AR	Caucasian/European	1 in 88	98%	1 in 4,351	1 in 1,531,552
Pendred syndrome	SLC26A4	AR	East Asian	1 in 74	98%	1 in 3,651	1 in 1,080,696
Permanent neonatal diabetes mellitus	KCNJ11	AR	General	1 in 423	99%	1 in 42,201	<1 in 10 million
Permanent neonatal diabetes mellitus	KCNJ11	AR	Caucasian/European	1 in 232	99%	1 in 23,101	<1 in 10 million
Peroxisomal acyl-CoA oxidase deficiency	ACOX1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Phenylalanine hydroxylase deficiency (Phenylketonuria)	PAH	AR	General	1 in 93	99%	1 in 9,201	1 in 3,422,772
Phenylalanine hydroxylase deficiency (Phenylketonuria)	PAH	AR	Caucasian/European	1 in 63	99%	1 in 6,201	1 in 1,562,652
Phenylalanine hydroxylase deficiency (Phenylketonuria)	PAH	AR	Middle-Eastern	1 in 74	99%	1 in 7,301	1 in 2,161,096
Phenylalanine hydroxylase deficiency (Phenylketonuria)	PAH	AR	South East Asian	1 in 59	99%	1 in 5,801	1 in 1,369,036
Phosphoglycerate dehydrogenase deficiency	PHGDH	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Phosphoglycerate dehydrogenase deficiency	PHGDH	AR	Ashkenazi Jewish	1 in 280	98%	1 in 13,951	<1 in 10 million
Phosphoglycerate kinase 1 deficiency	PGK1	AR	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
Phosphoribosylpyrophosphate synthetase superactivity	PRPS1	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
POLG-related disorders	POLG	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
Polycystic kidney disease, PKHD1-related	PKHD1	AR	General	1 in 70	98%	1 in 3,451	1 in 966,280
Polycystic kidney disease, PKHD1-related	PKHD1	AR	Ashkenazi Jewish	1 in 107	98%	1 in 5,301	1 in 2,268,828
Pompe disease	GAA	AR	General	1 in 100	98%	1 in 4,951	1 in 1,980,400

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Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
Pompe disease	GAA	AR	African/African American	1 in 60	98%	1 in 2,951	1 in 708,240
Pompe disease	GAA	AR	East Asian	1 in 112	98%	1 in 5,551	1 in 2,486,848
Pontocerebellar hypoplasia, type 1A	VRK1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Pontocerebellar hypoplasia, type 1B	EXOSC3	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Pontocerebellar hypoplasia, type 2D	SEPSECS	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Pontocerebellar hypoplasia, type 6	RARS2	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Postnatal progressive microcephaly with seizures and brain atrophy	MED17	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Primary ciliary dyskinesia, DNAH5-related	DNAH5	AR	General	1 in 142	98%	1 in 7,051	1 in 4,004,968
Primary ciliary dyskinesia, DNAI1-related	DNAI1	AR	General	1 in 230	98%	1 in 11,451	<1 in 10 million
Primary ciliary dyskinesia, DNAI2-related	DNAI2	AR	General	1 in 447	98%	1 in 22,301	<1 in 10 million
Primary ciliary dyskinesia, DNAL1-related	DNAL1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Primary ciliary dyskinesia, type 14	CCDC39	AR	General	1 in 211	98%	1 in 10,501	1 in 8,862,844
Primary ciliary dyskinesia, type 17	CCDC103	AR	General	1 in 316	98%	1 in 15,751	<1 in 10 million
Primary ciliary dyskinesia, type 30	CCDC151	AR	General	1 in 365	98%	1 in 18,201	<1 in 10 million
Primary congenital glaucoma	CYP1B1	AR	General	1 in 50	99%	1 in 4,901	1 in 980,200
Primary hyperoxaluria, type 1	AGXT	AR	General	1 in 120	99%	1 in 11,901	1 in 5,712,480
Primary hyperoxaluria, type 1	AGXT	AR	Caucasian/European	1 in 173	99%	1 in 17,201	<1 in 10 million
Primary hyperoxaluria, type 2	GRHPR	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Primary hyperoxaluria, type 3	HOGA1	AR	General	1 in 184	99%	1 in 18,301	<1 in 10 million
Progressive external ophthalmoplegia	POLG	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
Progressive familial intrahepatic cholestasis	ABCB11	AR	General	1 in 112	98%	1 in 5,551	1 in 2,486,848
Propionic acidaemia, PCCA-related	PCCA	AR	General	1 in 224	96%	1 in 5,576	1 in 4,996,096
Propionic acidaemia, PCCA-related	PCCA	AR	Native American	1 in 85	96%	1 in 2,101	1 in 714,340
Propionic acidaemia, PCCB-related	PCCB	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
Propionic acidaemia, PCCB-related	PCCB	AR	Native American	1 in 85	99%	1 in 8,401	1 in 2,856,340
Pycnodysostosis	CTSK	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Pyruvate carboxylase deficiency	PC	AR	General	1 in 250	95%	1 in 4,981	1 in 4,981,000
Pyruvate dehydrogenase E1-alpha deficiency	PDHA1	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
Pyruvate dehydrogenase E1-beta deficiency	PDHB	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Renal tubular acidosis with deafness	ATP6V1B1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Renpenning syndrome	PQBP1	XL	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Retinitis pigmentosa 12	CRB1	AR	General	1 in 104	98%	1 in 5,151	1 in 2,142,816
Retinitis pigmentosa 20	RPE65	AR	General	1 in 228	98%	1 in 11,351	<1 in 10 million
Retinitis pigmentosa 25	EYS	AR	General	1 in 66	98%	1 in 3,251	1 in 858,264
Retinitis pigmentosa 26	CERKL	AR	General	1 in 148	98%	1 in 7,351	1 in 4,351,792
Retinitis pigmentosa 28	FAM161A	AR	General	1 in 296	98%	1 in 14,751	<1 in 10 million
Retinitis pigmentosa 37	NR2E3	AR	General	1 in 209	98%	1 in 10,401	1 in 8,695,236
Retinitis pigmentosa 59	DHDDS	AR	General	1 in 296	98%	1 in 14,751	<1 in 10 million
Retinitis pigmentosa 59	DHDDS	AR	Ashkenazi Jewish	1 in 118	98%	1 in 5,851	1 in 2,761,672
Retinitis pigmentosa 74	BBS2	AR	General	1 in 621	99%	1 in 62,001	<1 in 10 million
Retinitis pigmentosa 74	BBS2	AR	Ashkenazi Jewish	1 in 107	99%	1 in 10,601	1 in 4,537,228
Retinitis pigmentosa 76	POMGNT1	AR	General	1 in 462	98%	1 in 23,051	<1 in 10 million
Retinitis pigmentosa 76	POMGNT1	AR	Finnish	1 in 111	98%	1 in 5,501	1 in 2,442,444
Retinitis pigmentosa, CNGA1-related	CNGA1	AR	General	1 in 210	99%	1 in 20,901	<1 in 10 million
Retinitis pigmentosa, CNGB1-related	CNGB1	AR	General	1 in 296	99%	1 in 29,501	<1 in 10 million
Retinitis pigmentosa, IDH3B-related	IDH3B	AR	General	1 in 296	99%	1 in 29,501	<1 in 10 million
Retinitis pigmentosa, PDE6A-related	PDE6A	AR	General	1 in 133	99%	1 in 13,201	1 in 7,022,932
Rhizomelic chondrodysplasia punctata, type 1	PEX7	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
Rhizomelic chondrodysplasia punctata, type 3	AGPS	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Roberts syndrome	ESCO2	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Rosenberg-Chutorian syndrome	PRPS1	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
Sandhoff disease	HEXB	AR	General	1 in 600	98%	1 in 29,951	<1 in 10 million
Schimke immunoosseous dysplasia	SMARCAL1	AR	General	1 in 500	90%	1 in 4,991	1 in 9,982,000
Schopf-Schulz-Passarge syndrome	WNT10A	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Segawa syndrome	TH	AR	General	1 in 224	98%	1 in 11,151	1 in 9,991,296
Senior-Løken syndrome 1	NPHP1	AR	General	1 in 480	98%	1 in 23,951	<1 in 10 million
Senior-Løken syndrome 1	NPHP1	AR	Finnish	1 in 124	98%	1 in 6,151	1 in 3,050,896
Senior-Løken syndrome 6	CEP290	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
Severe combined immunodeficiency with sensitivity to ionising radiation	DCLRE1C	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Severe combined immunodeficiency, JAK3-related	JAK3	AR	General	1 in 299	99%	1 in 29,801	<1 in 10 million
Severe combined immunodeficiency, X-linked	IL2RG	XL	General	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
Severe congenital neutropaenia, HAX1-related	HAX1	AR	General	1 in 224	98%	1 in 11,151	1 in 9,991,296

AR autosomal recessive; XL X-linked.
*If patient not shown to be a carrier. ^ If patient not identified as a carrier and partner not tested for AR conditions

Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
Severe congenital neutropaenia, VPS45-related	VPS45	AR	General	1 in 224	98%	1 in 11,151	1 in 9,991,296
Severe congenital neutropaenia, WAS-related	WAS	XL	General	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
Short branched-chain acyl-CoA dehydrogenase (SBCAD) deficiency	ACADSB	AR	General	1 in 368	99%	1 in 36,701	<1 in 10 million
Short branched-chain acyl-CoA dehydrogenase (SBCAD) deficiency	ACADSB	AR	Hmong	1 in 6	99%	1 in 501	<1 in 10 million
Short branched-chain acyl-CoA dehydrogenase (SBCAD) deficiency	ACADSB	AR	General	1 in 118	93%	1 in 1,672	1 in 789,184
Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	ACADS	AR	General	1 in 85	99%	1 in 8,401	1 in 2,856,340
Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	ACADS	AR	African/African American	1 in 52	99%	1 in 5,101	1 in 1,061,008
Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	ACADS	AR	Caucasian/European	1 in 76	99%	1 in 7,501	1 in 2,280,304
Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	ACADS	AR	Middle-Eastern	1 in 52	99%	1 in 5,101	1 in 1,061,008
Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	ACADS	AR	South Asian/Indian	1 in 51	99%	1 in 5,001	1 in 1,020,204
Sialic acid storage disorder	SLC17A5	AR	General	<1 in 500	91%	1 in 5,545	<1 in 10 million
Sialic acid storage disorder	SLC17A5	AR	Finnish	1 in 100	91%	1 in 1,101	1 in 440,400
Sickle cell disease	HBB	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
Sickle cell disease	HBB	AR	African/African American	1 in 10	95%	1 in 181	1 in 7,240
Sickle cell disease	HBB	AR	East Asian	1 in 50	95%	1 in 981	1 in 196,200
Sickle cell disease	HBB	AR	Latino	1 in 128	95%	1 in 2,541	1 in 1,300,992
Sickle cell disease	HBB	AR	Mediterranean	1 in 3	95%	1 in 41	1 in 492
Sickle cell disease	HBB	AR	South Asian/Indian	1 in 25	95%	1 in 481	1 in 48,100
Sjögren-Larsson syndrome	ALDH3A2	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
Smith-Lemli-Opitz syndrome	DHCR7	AR	General	1 in 30	96%	1 in 726	1 in 87,120
Smith-Lemli-Opitz syndrome	DHCR7	AR	African/African American	1 in 138	96%	1 in 3,426	1 in 1,891,152
Smith-Lemli-Opitz syndrome	DHCR7	AR	Ashkenazi Jewish	1 in 36	96%	1 in 876	1 in 126,144
Spastic paraplegia, type 15	ZFYVE26	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Spastic paraplegia, type 49	TECPR2	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Spastic paraplegia, type 7	SPG7	AR	General	1 in 159	99%	1 in 15,801	<1 in 10 million
SPG11-related neuromuscular disorders	SPG11	AR	General	1 in 159	99%	1 in 15,801	<1 in 10 million
Spinal muscular atrophy	SMN1	AR	General	1 in 54	91%	1 in 590	1 in 127,440
Spinal muscular atrophy	SMN1	AR	African/African American	1 in 72	71%	1 in 246	1 in 70,848
Spinal muscular atrophy	SMN1	AR	Ashkenazi Jewish	1 in 67	91%	1 in 734	1 in 196,712
Spinal muscular atrophy	SMN1	AR	Caucasian/European	1 in 47	95%	1 in 921	1 in 173,148
Spinal muscular atrophy	SMN1	AR	East Asian	1 in 59	93%	1 in 830	1 in 195,880
Spinal muscular atrophy	SMN1	AR	Latino	1 in 68	90%	1 in 671	1 in 182,512
Spondylocostal dysostosis	MESP2	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Steel syndrome	COL27A1	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Stüve-Wiedemann syndrome	LIFR	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Systemic primary carnitine deficiency	SLC22A5	AR	General	1 in 129	76%	1 in 534	1 in 275,544
Systemic primary carnitine deficiency	SLC22A5	AR	African/African American	1 in 86	76%	1 in 355	1 in 122,120
Systemic primary carnitine deficiency	SLC22A5	AR	East Asian	1 in 77	76%	1 in 318	1 in 97,944
Systemic primary carnitine deficiency	SLC22A5	AR	Faroese	1 in 9	76%	1 in 34	1 in 1,224
Systemic primary carnitine deficiency	SLC22A5	AR	Pacific Islander	1 in 37	76%	1 in 151	1 in 22,348
Systemic primary carnitine deficiency	SLC22A5	AR	South Asian/Indian	1 in 51	76%	1 in 209	1 in 42,636
Tay-Sachs disease	HEXA	AR	General	1 in 300	99%	1 in 29,901	<1 in 10 million
Tay-Sachs disease	HEXA	AR	Ashkenazi Jewish	1 in 27	99%	1 in 2,601	1 in 280,908
Tetrahydrobiopterin deficiency	PTS	AR	General	1 in 354	96%	1 in 8,826	<1 in 10 million
Tetrahydrobiopterin deficiency, PCBD1-related	PCBD1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Tetrahydrobiopterin deficiency, QDPR-related	QDPR	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Thrombocytopaenia, X-linked	WAS	XL	General	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
Thyroid dyshormonogenesis, IYD-related	IYD	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Thyroid dyshormonogenesis, SLC5A5-related	SLC5A5	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Thyroid dyshormonogenesis, TG-related	TG	AR	General	1 in 241	99%	1 in 24,001	<1 in 10 million
Thyroid dyshormonogenesis, TPO-related	TPO	AR	General	1 in 373	99%	1 in 37,201	<1 in 10 million
Treacher Collins syndrome, POLR1C-related	POLR1C	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
Trichohepatoenteric syndrome	TTC37	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Trifunctional protein deficiency	HADHA	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Trifunctional protein deficiency	HADHA	AR	Finnish	1 in 124	98%	1 in 6,151	1 in 3,050,896
Tyrosinaemia, type 1	FAH	AR	General	1 in 99	95%	1 in 1,961	1 in 776,556
Tyrosinaemia, type 1	FAH	AR	Ashkenazi Jewish	1 in 150	95%	1 in 2,981	1 in 1,788,600

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Correct at time of print | September 2019

Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
Tyrosinaemia, type 1	FAH	AR	Finnish	1 in 122	95%	1 in 2,421	1 in 1,181,448
Tyrosinaemia, type 1	FAH	AR	French Canadian	1 in 66	95%	1 in 1,301	1 in 343,464
Tyrosinaemia, type 1	FAH	AR	South Asian/Indian	1 in 172	95%	1 in 3,421	1 in 2,353,648
Tyrosinaemia, type 2	TAT	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
Usher syndrome, type 1B	MYO7A	AR	General	1 in 206	98%	1 in 10,251	1 in 8,446,824
Usher syndrome, type 1B	MYO7A	AR	East Asian	1 in 62	98%	1 in 3,051	1 in 756,648
Usher syndrome, type 1C	USH1C	AR	General	1 in 353	90%	1 in 3,521	1 in 4,971,652
Usher syndrome, type 1C	USH1C	AR	French Canadian	1 in 227	90%	1 in 2,261	1 in 2,052,988
Usher syndrome, type 1D	CDH23	AR	General	1 in 285	90%	1 in 2,841	1 in 11,364
Usher syndrome, type 1F	PCDH15	AR	General	1 in 395	98%	1 in 19,701	1 in 78,804
Usher syndrome, type 1F	PCDH15	AR	Ashkenazi Jewish	1 in 72	98%	1 in 3,551	1 in 14,204
Usher syndrome, type 1G	USH1G	AR	General	1 in 434	99%	1 in 43,301	<1 in 10 million
Usher syndrome, type 2A	USH2A	AR	General	1 in 126	96%	1 in 3,126	1 in 1,575,504
Usher syndrome, type 2A	USH2A	AR	Caucasian/European	1 in 73	96%	1 in 1,801	1 in 525,892
Usher syndrome, type 2D	WHRN	AR	General	1 in 282	99%	1 in 28,101	<1 in 10 million
Usher syndrome, type 3A	CLRN1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
Usher syndrome, type 3A	CLRN1	AR	Ashkenazi Jewish	1 in 120	98%	1 in 5,951	1 in 2,856,480
Usher syndrome, type 3A	CLRN1	AR	Finnish	1 in 70	98%	1 in 3,451	1 in 966,280
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL	AR	Middle-Eastern	1 in 74	93%	1 in 1,044	1 in 309,024
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL	AR	Native American	1 in 61	93%	1 in 858	1 in 209,352
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL	AR	South Asian/Indian	1 in 73	93%	1 in 1,030	1 in 300,760
Weyers acrodental dysostosis, EVC2-related	EVC2	AR	General	1 in 240	98%	1 in 11,951	<1 in 10 million
Weyers acrodental dysostosis, EVC2-related	EVC2	AR	Amish	1 in 7	98%	1 in 301	1 in 8,428
Weyers acrofacial dysostosis, EVC-related	EVC	AR	General	1 in 142	98%	1 in 7,051	1 in 4,004,968
Weyers acrofacial dysostosis, EVC-related	EVC	AR	Amish	1 in 7	98%	1 in 301	1 in 8,428
Wilson disease	ATP7B	AR	General	1 in 87	98%	1 in 4,301	1 in 1,496,748
Wilson disease	ATP7B	AR	Caucasian/European	1 in 42	98%	1 in 2,051	1 in 344,568
Wilson disease	ATP7B	AR	Ashkenazi Jewish	1 in 70	98%	1 in 3,451	1 in 966,280
Wiskott-Aldrich syndrome	WAS	XL	General	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
Wolcott-Rallison syndrome	EIF2AK3	AR	General	<1 in 500	98%	1 in 24,951	<1 in 10 million
Xeroderma pigmentosum, group A	XPA	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
Xeroderma pigmentosum, group A	XPA	AR	Japanese	1 in 74	99%	1 in 7,301	1 in 2,161,096
Xeroderma pigmentosum, group C	XPC	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
X-linked Aarskog-Scott syndrome	FGD1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked epilepsy with variable learning disabilities	SYN1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked hearing loss, POU3F4-related	POU3F4	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability with cerebellar hypoplasia and distinctive facial appearance	OPHN1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability, AP1S2-related	AP1S2	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability, ARX-related	ARX	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability, BRWD3-related	BRWD3	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability, CUL4B-related	CUL4B	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability, DLG3-related	DLG3	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability, FTSJ1-related	FTSJ1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability, IL1RAPL1-related	IL1RAPL1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability, KDM5C-related	KDM5C	XL	General	<1 in 50,000	98%	1 in 2,499,951	<1 in 10 million
X-linked intellectual disability, PAK3-related	PAK3	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability, Siderius type	PHF8	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million

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Expanded carrier screen

Gene list (by disease)

Disorder	Gene	Inheritance	Population	Carrier frequency	Detection rate	Residual risk of being a carrier*	Residual risk of having an affected child^
X-linked intellectual disability, THOC2-related	THOC2	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
X-linked intellectual disability, ZNF711-related	ZNF711	XL	General	<1 in 50,000	93%	1 in 714,272	1 in 2,857,143
X-linked ocular albinism, GPR143-related	GPR143	XL	General	1 in 25,000	99%	1 in 2,499,901	<1 in 10 million
X-linked retinitis pigmentosa, RP2-related	RP2	XL	General	1 in 4,000	99%	1 in 399,901	1 in 1,600,000
X-linked retinitis pigmentosa, RPGR-related	RPGR	XL	General	1 in 3,000	75%	1 in 11,997	1 in 48,000
Zellweger syndrome, PEX10-related	PEX10	AR	General	1 in 500	95%	1 in 9,981	<1 in 10 million
Zellweger syndrome, PEX10-related	PEX10	AR	Japanese	1 in 354	95%	1 in 7,061	1 in 9,998,376
Zellweger syndrome, PEX12-related	PEX12	AR	General	1 in 373	95%	1 in 7,441	<1 in 10 million
Zellweger syndrome, PEX1-related	PEX1	AR	General	1 in 147	95%	1 in 2,921	1 in 1,717,548
Zellweger syndrome, PEX2-related	PEX2	AR	General	1 in 500	95%	1 in 9,981	<1 in 10 million
Zellweger syndrome, PEX2-related	PEX2	AR	Ashkenazi Jewish	1 in 123	95%	1 in 2,441	1 in 1,200,972
Zellweger syndrome, PEX6-related	PEX6	AR	General	1 in 280	95%	1 in 5,581	1 in 6,250,720

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