

## Test by disease

CONDITION(S)	GENE	ETHNIC BACKGROUND	DETECTION RATE	CARRIER RISK	RESIDUAL RISK
3-Hydroxy-3-methylglutaric aciduria, HMG-CoA lyase deficiency	HMGCL	Saudi Arabian	87%	1 in 158	1 in 1209
		Spanish / Portugese	97%	1 in 158	1 in 5234
Andermann Syndrome (Agenesis of the Corpus Colosum with Peripheral Neuropathy)	SLC12A6	French Canadian	99%	1 in 23	1 in 2201
		General	48%	1 in 76	1 in 145
Argininosuccinate aciduria	ASL	General	30%	1 in 132	1 in 188
Aspartylglucosaminuria	AGA	Finnish	98%	1 in 68	1 in 3351
Ataxia Telangiectasia	ATM	Amish	99%	1 in 100	1 in 9901
		N. African Jews	97%	1 in 100	1 in 3234
		General	24%	1 in 100	1 in 133
Bardet-Biedl syndrome 1	BBS1	General	80%	<1 in 187	<1 in 931
Batten Disease, Juvenile Neuronal Ceroid Lipofuscinosis	CLN3	Icelandic	81%	1 in 79	1 in 412
Biotinidase Deficiency; Multiple Carboxylase Deficiency	BTM	General	52%	1 in 186	1 in 387
Bloom Syndrome	BLM	Ashkenazi	97%	1 in 107	1 in 3534
Carnitin Palmitoyltransferase II Deficiency	CPT2	General	90%	<1 in 500	1 in 4991
Carnitine Uptake Defect	SLC22A5	Chinese	83%	1 in 125	1 in 730
Canavan	ASPA	Ashkenazi	99%	1 in 41	1 in 2001
		General	60%	<1 in 100	<1 in 250
Cartilage-Hair Hypoplasia	RMRP	Finnish	92%	1 in 76	1 in 938
Ceroid Lipofuscinosis, Neuronal 2; CLN2	TPP1	Caucasian	53%	1 in 158	1 in 337
Ceroid Lipofuscinosis, Neuronal 1; CLN1	PPT1	Finnish	98%	<1 in 80	1 in 3951
		General	75%	<1 in 80	<1 in 159
Citrullinemia type I	ASS1	Japanese	75%	1 in 120	1 in 477
Congenital Disorder of Glycosylation-1a	PMM2	Dutch/Danish	88%	1 in 71	1 in 584
Creatine transporter defect, X-linked MR with seizures, short stature, and midface hypoplasia	SLC6A8	General	20%	1 in 8500 females	1 in 10,625 females
Cystic Fibrosis	CFTR	Caucasian	91%	1 in 27	1 in 290
		Ashkenazi	96%	1 in 27	1 in 651
		Hispanic	81%	1 in 46	1 in 156
		African American	78%	1 in 65	1 in 292
		Asian American	49%	1 in 94	1 in 182
Diastrophic Dysplasia	SLC26A2	General	65%	1 in 158	1 in 452
		Finnish	96%	1 in 50	1 in 1226
Dihydropyrimidinase Deficiency	DLD	Ashkenazi	95%	1 in 110	1 in 2180
Familial Dysautonomia	IKBKAP	Ashkenazi	99%	1 in 32	1 in 3101
Familial Mediterranean Fever	MEFV	Turkish	51%	1 in 5	1 in 9
		Syrian	89%	1 in 6	1 in 46
		Armenian	90%	1 in 6	1 in 51
Fanconi Type C	FANCC	Ashkenazi	99%	1 in 89	1 in 8801

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Galactokinase Deficiency, Galactosemia II, GALK deficiency	GALK1	Romani (gypsies)	99%	1 in 47	1 in 4601
		Japanese	60%	1 in 500	1 in 1248
Galactosemia	GALT	General	75%	1 in 122	1 in 485
Glutaric Acidemia type 1 (GA-1)	GCDH,	Amish	99%	1 in 94	1 in 9301
		German	40%	1 in 94	1 in 156
Glycogen Storage Disease type 1a	G6PC	Ashkenazi	99%	1 in 71	1 in 7001
		General	69%	1 in 158	1 in 507
Glycogen Storage Disease type 1b	SLC37A4	General	50%	1 in 112	1 in 223
GRACILE syndrome	BCS1L	Finnish	99%	1 in 120	1 in 11,901
Guanidinoacetate Methyltransferase Deficiency	GAMT	Portuguese	80%	1 in 125	1 in 621
		General	33%	1 in 246	1 in 372
Hereditary Fructose Intolerance, Fructosemia	ALDOB	General	85%	1 in 71	1 in 468
Holocarboxylase Synthetase Deficiency	HLCS	General	63%	1 in 148	1 in 398
Homocystinuria	CBS	General	50%	1 in 112	1 in 223
Hurler, Scheie, Hurler-Scheie disease (MPS1)	IDUA	European	72%	1 in 160	1 in 569
		Scandinavian	79%	1 in 160	1 in 590
		Moroccan	92%	1 in 160	1 in 1989
		Italian	50%	1 in 160	1 in 319
		Korean	50%	1 in 160	1 in 319
Hyperoxaluria type 1	AGXT	General	74%	1 in 289	1 in 1109
Hyperoxaluria type 2	GRHPR	Asian	50%	<1 in 150	<1 in 210
Hyperornithinemia-Hyperammonemia- Homocitrullinuria Syndrome (HHH syndrome)	SLC25A15	French Canadian	95%	<1 in 500	<1 in 10,000
Hypophosphatasia, AR	ALPL	Caucasian/European	>35%	1 in 150	<1 in 230
		Mennonite	>90%	1 in 25	1 in 241
Joubert Syndrome 2	TMEM216	Ashkenazi	99%	1 in 92	1 in 9101
Krabbe disease, GALC Deficiency	GALC	General	66%	1 in 150	1 in 439
Leigh Syndrome, French-Canadian Type	LRPPRC	French Canadian	98%	1 in 22	1 in 1051
Long-chain L-3- hydroxyacyl-CoA dehydrogenase (LCHAD)	HADHA	General	87%	1 in 125	1 in 955
Maple Syrup Urine Disease	BCKDHA	Mennonite	99%	1 in 12	1 in 1101
		Ashkenazi	95%	1 in 80	1 in 1581
Medium-chain acyl-coenzyme A dehydrogenase (MCAD)	ACADM	General	90%	1 in 50	1 in 491
Methylmelonic Aciduria and Homocystinuria, cblc Type	MMACHC	General	67%	1 in 141	1 in 442
Methylmalonic Aciduria - MUT related	MUT	African American	41%	1 in 141	1 in 287
		Caucasian	20%	1 in 141	1 in 176
		Hispanic	60%	1 in 141	1 in 234
Methylmalonic Aciduria (cobalamin deficiency) cblA type	MMAA	General	43%	1 in 141	1 in 247
Methylmalonic Aciduria (cobalamin deficiency) cblB type	MMAB	General	58%	1 in 141	1 in 334
Mucopolipidosis type IV	MCOLN1	Ashkenazi	95%	1 in 100	1 in 1981

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Muscle-Eye-Brain Disease	POMGNT1	Finnish	96%	1 in 112	1 in 2776
Nemaline Myopathy	NEB	Ashkenazi	99%	1 in 168	1 in 6701
Nephrosis 1, congenital, Finnish type	NPHS1	Finnish	81%	1 in 45	1 in 233
		Maltese	99%	1 in 113	1 in 11,201
		General	31%	1 in 113	1 in 163
Nieman Pick Type A	SMPD1	Ashkenazi	90%	1 in 90	1 in 891
Nieman Pick Type B	SMPD1	North African	90%	1 in 250	1 in 2491
Nijmegen Breakage Syndrome	NBN	General	70%	1 in 158	1 in 524
Nonketotic hyperglycinemia (NKH); GLYCINE DECARBOXYLASE	GLDC	Finnish	78%	1 in 55	1 in 246
Ornithine Transcarbamylase Deficiency	OTC	General	14%	1 in 142	1 in 165
Phenylketonuria (PKU)	PAH	Caucasian	48%	1 in 50	1 in 93
Polycystic kidney and hepatic disease; Congenital Hepatic Fibrosis; Caroli Disease	PKHD1	General	39%	1 in 70	1 in 114
Pompe disease (Glycogen storage disease type II)	GAA	Dutch	67%	1 in 100	1 in 301
		Italian	42%	1 in 100	1 in 172
		Chinese	85%	1 in 100	1 in 661
		African American	60%	1 in 100	1 in 249
Progressive Epilepsy with Mental Retardation (EPMR)	CLN8	Finnish	99%	1 in 210	1 in 20,801
Recessive multiple epiphyseal dysplasia	SLC26A2	General	65%	1 in 71	1 in 701
Rhizomelic Chondrodysplasia Punctata Type 1	PEX7	General	68%	1 in 167	1 in 520
Salla Disease	SLC17A5	Finnish	95%	1 in 40	1 in 976
Severe Combined Immunodeficiency (AR), Adenosine deaminase deficiency	ADA	African - Caribbean	64%	1 in 224	1 in 620
Sjogren-Larsson syndrome	FALDH	Swedish	83%	1 in 50	1 in 328
		General	56%	1 in 250	1 in 567
Smith-Lemli-Opitz Syndrome	DHCR7	General	70%	1 in 94	1 in 311
Spastic Ataxia of Charlevoix-Saguenay	SACS	French Canadian	97%	1 in 22	1 in 701
Steroid-Resistant Nephrotic Syndrome	NPHS2	General	73%	1 in 400	1 in 1479
Tay-Sachs Disease	HEXA	Ashkenazi	96%	1 in 28	1 in 696
		French Canadian	83%	1 in 65	1 in 377
Tyrosinemia Type 1	FAH	French Canadian	88%	1 in 66	1 in 543
		European	54%	1 in 112	1 in 242
		Ashkenazi	99%	1 in 100	1 in 9901
Usher Syndrome Type III	CLRN1	Ashkenazi	98%	1 in 120	1 in 5951
Usher Syndrome Type 1F	PCDH15	Ashkenazi	75%	1 in 147	1 in 585
Variant late infantile Neuronal Ceroid Lipofuscinosis CLN5	CLN5	Finnish	94%	<1 in 80	1 in 1318
X-linked Severe Combined Immunodeficiency	IL2RG	General	21%	1 in 112 in females	1 in 141 in females
Zellweger, Neonatal Adrenoleukodystrophy, Infantile Refsum disease	PEX1	General	81%	1 in 112	1 in 585