Gene	Disorder	Ethnicity	Detection Rate (%)	Individual Carrier Risk (a priori)	Individual Residual Risk After Negative Result	Risk of Affected Fetus (Carrier & Negative Partners Couple)
AAAS	Triple A syndrome	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 422	1 in 42100	1 in 168400
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
ABCA12	Autosomal recessive congenital ichthyosis (ARCI)	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	1 in 423	1 in 21100	1 in 84400
		General population	99%	1 in 325	1 in 32400	1 in 129600
ABCA4	Stargardt disease	General population	98%	1 in 51	1 in 2500	1 in 10000
ABCB11	Progressive familial intrahepatic cholestasis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 350	1 in 34900	1 in 139600
		General population	99%	1 in 374	1 in 37300	1 in 149200
ABCB4	Progressive familial intrahepatic cholestasis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 467	1 in 46600	1 in 186400
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
ABCC6	Pseudoxanthoma elasticum	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
ABCC8	Familial hyperinsulinism	Ashkenazi Jewish	99%	1 in 74	1 in 7300	1 in 29200
		Finnish	99%	1 in 224	1 in 22300	1 in 89200
		Non-Finnish European/White	99%	1 in 297	1 in 29600	1 in 118400
		General population	99%	1 in 491	1 in 49000	1 in 196000
ABCD1	Adrenoleukodystrophy, X-linked	General population (Female)	99%	~1 in 20000	N/A	N/A
ACAD9	Mitochondrial complex I deficiency	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	Ashkenazi Jewish	99%	1 in 114	1 in 11300	1 in 45200
		Finnish	99%	1 in 399	1 in 39800	1 in 159200
		Non-Finnish European/White	99%	1 in 56	1 in 5500	1 in 22000
		General population	99%	1 in 67	1 in 6600	1 in 26400
ACADS	Short Chain Acyl-CoA Dehydrogenase (SCAD) deficiency	Finnish	99%	<1 in 500	<1 in 49900	<1 in 199600
		Ashkenazi Jewish	99%	1 in 20	1 in 1900	1 in 7600
ACADSB	Short/branched chain acyl-CoA dehydrogenase deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 478	1 in 47700	1 in 190800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 261	1 in 26000	1 in 104000

		Non-Finnish European/White	99%	1 in 122	1 in 12100	1 in 48400
		General population	99%	1 in 118	1 in 11700	1 in 46800
ACAT1	Beta-ketothiolase deficiency	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	1 in 431	1 in 43000	1 in 172000
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
ACSF3	Combined malonic and methylmalonic aciduria	Ashkenazi Jewish	99%	1 in 341	1 in 34000	1 in 136000
		Finnish	99%	1 in 331	1 in 33000	1 in 132000
		Non-Finnish European/White	99%	1 in 71	1 in 7000	1 in 28000
		General population	99%	1 in 121	1 in 12000	1 in 48000
ADA	Adenosine deaminase deficiency	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	99%	1 in 437	1 in 43600	1 in 174400
		General population	99%	1 in 390	1 in 38900	1 in 155600
ADAMTS2	Ehlers Danlos syndrome, ADAMTS2-related	Ashkenazi Jewish	99%	1 in 168	1 in 16700	1 in 66800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	97%	< 1 in 500	<1 in 16630	<1 in 66520
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ADGRG1	Polymicrogyria	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
AGA	Aspartylglucosaminuria	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	1 in 61	1 in 6000	1 in 24000
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	1 in 264	1 in 26300	1 in 105200
AGL	Glycogen storage disease type III	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 260	1 in 25900	1 in 103600
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
AGPS	Rhizomelic chondrodysplasia punctata	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	97%	< 1 in 500	<1 in 16630	<1 in 66520
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
AGXT	Primary hyperoxaluria	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 235	1 in 23400	1 in 93600
		General population	99%	1 in 257	1 in 25600	1 in 102400
AIRE	Autoimmune polyglandular syndrome type 1	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	1 in 93	1 in 9200	1 in 36800

		Non-Finnish European/White	98%	1 in 207	1 in 10300	1 in 41200
		General population	98%	1 in 181	1 in 9000	1 in 36000
ALDH3A2	Sjogren-Larsson syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ALDH7A1	Pyridoxine-dependent epilepsy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 326	1 in 32500	1 in 130000
		General population	99%	1 in 375	1 in 37400	1 in 149600
ALDOB	Hereditary fructose Intolerance	Ashkenazi Jewish	99%	1 in 137	1 in 13600	1 in 54400
		Finnish	99%	1 in 102	1 in 10100	1 in 40400
		Non-Finnish European/White	99%	1 in 82	1 in 8100	1 in 32400
		General population	99%	1 in 112	1 in 11100	1 in 44400
ALG6	Congenital disorders of glycosylation	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 330	1 in 32900	1 in 131600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ALMS1	Alström syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	1 in 151	1 in 7500	1 in 30000
		General population	98%	1 in 334	1 in 16650	1 in 66600
ALPL	Hypophosphatasia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 30	1 in 2900	1 in 11600
		Non-Finnish European/White	99%	1 in 139	1 in 13800	1 in 55200
		General population	99%	1 in 128	1 in 12700	1 in 50800
AMH	Persistent Müllerian duct syndrome, type I	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
AMHR2	Persistent Müllerian duct syndrome, type II	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
AMT	Glycine encephalopathy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
AP1S1	MEDNIK syndrome	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
AQP2	Nephrogenic diabetes insipidus	Finnish	95%	<1 in 500	<1 in 9980	<1 in 39920
		General population	95%	1 in 169	1 in 3360	1 in 13440
AR	Androgen Insensitivity Syndrome (X-linked)	General population	99%	1 in 320	1 in 31900	1 in 127600
ARG1	Arginase deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ARSA	Metachromatic leukodystrophy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 236	1 in 23500	1 in 94000

		Non-Finnish European/White	99%	1 in 135	1 in 13400	1 in 53600
		General population	99%	1 in 152	1 in 15100	1 in 60400
ARSB	Mucopolysaccharidosis type VI	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 358	1 in 35700	1 in 142800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
ASL	Argininosuccinic aciduria	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 94	1 in 9300	1 in 37200
		Non-Finnish European/White	99%	1 in 142	1 in 14100	1 in 56400
		General population	99%	1 in 116	1 in 11500	1 in 46000
ASNS	Asparagine synthetase deficiency	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ASPA	Canavan disease	Ashkenazi Jewish	99%	1 in 48	1 in 4700	1 in 18800
		Finnish	99%	1 in 254	1 in 25300	1 in 101200
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	1 in 314	1 in 31300	1 in 125200
ASS1	Citrullinemia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 338	1 in 33700	1 in 134800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ATM	Ataxia-telangiectasia	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	1 in 155	1 in 7700	1 in 30800
		General population	98%	1 in 207	1 in 10300	1 in 41200
ATP6V1B1	Renal tubular acidosis with deafness	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ATP7A	ATP7A-related copper transport disorders (incl. Menkes disease)	General population (Female)	99%	~1 in 17000	N/A	N/A
ATP7B	Wilson disease	Ashkenazi Jewish	99%	1 in 42	1 in 4100	1 in 16400
		Finnish	99%	1 in 215	1 in 21400	1 in 85600
		Non-Finnish European/White	99%	1 in 72	1 in 7100	1 in 28400
		General population	99%	1 in 98	1 in 9700	1 in 38800
ATP8B1	Progressive familial intrahepatic cholestasis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ATRX	Alpha-thalassemia X-linked intellectual disability	General population (Female)	99%	~1 in 15000	N/A	N/A
BBS1	Bardet-Biedl syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600

		Finnish	99%	1 in 290	1 in 28900	1 in 115600
		Non-Finnish European/White	99%	1 in 149	1 in 14800	1 in 59200
		General population	99%	1 in 274	1 in 27300	1 in 109200
BBS10	Bardet-Biedl syndrome	Ashkenazi Jewish	97%	1 in 305	1 in 10130	1 in 40520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	97%	1 in 248	1 in 8230	1 in 32920
		General population	98%	1 in 438	1 in 21850	1 in 87400
BBS12	Bardet-Biedl syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
BBS2	Bardet-Biedl syndrome	Ashkenazi Jewish	99%	1 in 126	1 in 12500	1 in 50000
		Finnish	99%	1 in 499	1 in 49800	1 in 199200
		Non-Finnish European/White	99%	1 in 423	1 in 42200	1 in 168800
		General population	99%	1 in 479	1 in 47800	1 in 191200
BBS4	Bardet-Biedl syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
BBS9	Bardet-Biedl syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
BCKDHA	Maple syrup urine disease	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
BCKDHB	Maple syrup urine disease	Ashkenazi Jewish	99%	1 in 74	1 in 7300	1 in 29200
		Finnish	99%	1 in 175	1 in 17400	1 in 69600
		Non-Finnish European/White	99%	1 in 349	1 in 34800	1 in 139200
		General population	98%	1 in 378	1 in 18850	1 in 75400
BCS1L	GRACILE syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 99	1 in 9800	1 in 39200
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
BCHE	Pseudocholinesterase deficiency	Ashkenazi Jewish	99%	1 in 21	1 in 2000	1 in 8000
		Finnish	99%	1 in 36	1 in 3500	1 in 14000
		Non-Finnish European/White	99%	1 in 18	1 in 1700	1 in 6800
		General population	99%	1 in 21	1 in 2000	1 in 8000
BLM	Bloom syndrome	Ashkenazi Jewish	97%	1 in 123	1 in 4070	1 in 16280
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 327	1 in 32600	1 in 130400
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
BRIP1	Fanconi anemia	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600

		Non-Finnish European/White	98%	1 in 340	1 in 16950	1 in 67800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
BSND	Bartter syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
BTD	Biotinidase deficiency, profound and partial	Ashkenazi Jewish	99%	1 in 16	1 in 1500	1 in 6000
		Finnish	99%	1 in 10	1 in 900	1 in 3600
		Non-Finnish European/White	99%	1 in 12	1 in 1100	1 in 4400
		General population	99%	1 in 16	1 in 1500	1 in 6000
	Biotinidase deficiency, profound	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 168	1 in 16700	1 in 66800
		General population	99%	1 in 265	1 in 26400	1 in 105600
BTK	Agammaglobulinemia, X-linked	General population	99%	N/A	N/A	N/A
CANT1	Desbuquois dysplasia type 1	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
CAPN3	Limb-girdle muscular dystrophy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 167	1 in 16600	1 in 66400
		General population	99%	1 in 265	1 in 26400	1 in 105600
CASQ2	Catecholaminergic polymorphic VT type 2	General population	99%	1 in 224	1 in 22300	1 in 89200
CBS	Homocystinuria	Ashkenazi Jewish	99%	1 in 337	1 in 33600	1 in 134400
		Finnish	99%	1 in 455	1 in 45400	1 in 181600
		Non-Finnish European/White	99%	1 in 142	1 in 14100	1 in 56400
		General population	99%	1 in 211	1 in 21000	1 in 84000
CC2D1A	Non-syndromic intellectual disability 3	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
CCN6	Progressive pseudorheumatoid dysplasia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
CDH23	Usher syndrome (hearing loss and retinitis pigmentosa)	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 281	1 in 28000	1 in 112000
		General population	98%	1 in 331	1 in 16500	1 in 66000
CEP290	Ciliopathies	Ashkenazi Jewish	99%	1 in 440	1 in 43900	1 in 175600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	1 in 109	1 in 5400	1 in 21600
		General population	98%	1 in 169	1 in 8400	1 in 33600
CERKL	Retinitis pigmentosa	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 48	1 in 4700	1 in 18800
		Non-Finnish European/White	99%	1 in 361	1 in 36000	1 in 144000
		General population	99%	1 in 305	1 in 30400	1 in 121600

CFTR	Cystic fibrosis	Ashkenazi Jewish	99%	1 in 17	1 in 1600	1 in 6400
		Finnish	98%	1 in 73	1 in 3600	1 in 14400
		Non-Finnish European/White	98%	1 in 21	1 in 1000	1 in 4000
		General population	98%	1 in 33	1 in 1600	1 in 6400
CIITA	Bare lymphocyte syndrome type II	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
CLN3	Neuronal ceroid-lipofuscinosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
CLN5	Neuronal ceroid-lipofuscinosis	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
CLN6	Neuronal ceroid-lipofuscinosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
CLN8	Neuronal ceroid-lipofuscinosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 431	1 in 43000	1 in 172000
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
CLRN1	Usher syndrome (hearing loss and retinitis pigmentosa)	Ashkenazi Jewish	99%	1 in 87	1 in 8600	1 in 34400
		Finnish	99%	1 in 71	1 in 7000	1 in 28000
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	1 in 312	1 in 31100	1 in 124400
CNGA3	Achromatopsia 2	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
CNGB3	Achromatopsia	Ashkenazi Jewish	99%	1 in 272	1 in 27100	1 in 108400
		Finnish	97%	1 in 172	1 in 5700	1 in 22800
		Non-Finnish European/White	97%	1 in 119	1 in 3930	1 in 15720
		General population	98%	1 in 149	1 in 7400	1 in 29600
COL11A2	Otospondylomegaepiphyseal dysplasia	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
COL4A3	Alport syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
COL4A4	Alport syndrome	Ashkenazi Jewish	99%	1 in 178	1 in 17700	1 in 70800
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 360	1 in 35900	1 in 143600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
COL4A5	Alport syndrome	General population (Female)	99%	~1 in 5000	N/A	N/A
COL7A1	Dystrophic epidermolysis bullosa	Ashkenazi Jewish	98%	1 in 193	1 in 9600	1 in 38400

		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 163	1 in 16200	1 in 64800
		General population	98%	1 in 253	1 in 12600	1 in 50400
CPS1	Carbamoyl phosphate synthetase I deficiency	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
CPT1A	Carnitine palmitoyltransferase I deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 274	1 in 27300	1 in 109200
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	1 in 349	1 in 34800	1 in 139200
CPT2	Carnitine palmitoyltransferase II deficiency	Ashkenazi Jewish	99%	1 in 40	1 in 3900	1 in 15600
		Finnish	99%	1 in 242	1 in 24100	1 in 96400
		Non-Finnish European/White	99%	1 in 178	1 in 17700	1 in 70800
		General population	99%	1 in 169	1 in 16800	1 in 67200
CRB1	Leber Congenital Amaurosis (CRB1-related)	General population	99%	~1 in 11000	N/A	
CTNS	Cystinosis	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
CTSC	Papillon-Lefèvre syndrome	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
CTSD	Neuronal ceroid-lipofuscinosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
CTSK	Pycnodysostosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
CYBA	Chronic granulomatous disease	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
CYBB	Chronic granulomatous disease	General population	99%	N/A	N/A	N/A
CYP11B1	11-hydroxylase deficient CAH	Non-Finnish European/White	98%	1 in 150	1 in 7500	1 in 29000
CYP11B2	Aldosterone synthase deficiency	Non-Finnish European/White	99%	<1 in 500	<1 in 49900	<1 in 199600
CYP17A1	Congenital adrenal hyperplasia	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
CYP19A1	AROM deficiency	General population	98%	<1 in 500	<1 in 24950	<1 in 99800

CYP1B1	Primary congenital glaucoma	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	99%	1 in 201	1 in 20000	1 in 80000
		General population	99%	1 in 128	1 in 12700	1 in 50800
CYP21A2	Congenital adrenal hyperplasia	Ashkenazi Jewish	90%	1 in 40	1 in 390	1 in 1560
		Non-Finnish European/White	88%	1 in 70	1 in 580	1 in 2320
CYP27A1	Cerebrotendinous xanthomatosis	Ashkenazi Jewish	99%	1 in 300	1 in 29900	1 in 119600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 299	1 in 29800	1 in 119200
		General population	99%	1 in 293	1 in 29200	1 in 116800
CYP27B1	Vitamin D-dependent rickets	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	1 in 397	1 in 39600	1 in 158400
DBT	Maple syrup urine disease	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 342	1 in 34100	1 in 136400
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
DCLRE1C	Omenn syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
DDB2	Xeroderma pigmentosum	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
DHCR7	Smith-Lemli-Opitz syndrome	Ashkenazi Jewish	99%	1 in 40	1 in 3900	1 in 15600
		Finnish	99%	1 in 181	1 in 18000	1 in 72000
		Non-Finnish European/White	99%	1 in 51	1 in 5000	1 in 20000
		General population	99%	1 in 72	1 in 7100	1 in 28400
DHDDS	Retinitis pigmentosa	Ashkenazi Jewish	99%	1 in 97	1 in 9600	1 in 38400
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
DKC1	Dyskeratosis congenita, X-linked	General population	99%	N/A	N/A	N/A
DLD	Dihydrolipoamide dehydrogenase deficiency	Ashkenazi Jewish	99%	1 in 57	1 in 5600	1 in 22400
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
DMD	Dystrophinopathies, including Duchenne and Becker	General population (Female)	99%	~1 in 3000	N/A	N/A
DNAH5	Primary ciliary dyskinesia 3	General population	98%	1 in 142	1 in 7050	1 in 28200
DNAI1	Primary ciliary dyskinesia 1	General population	98%	1 in 230	1 in 11450	1 in 45800
DNAI2	Primary ciliary dyskinesia 9	General population	98%	1 in 447	1 in 22300	1 in 89200

DNAL1	Primary ciliary dyskinesia 16	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
DOK7	Congenital myasthenic syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	97%	1 in 281	1 in 9330	1 in 37320
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
DPYD	Dihydropyrimidine dehydrogenase deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 254	1 in 25300	1 in 101200
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
DYSF	Limb-girdle muscular dystrophy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 263	1 in 26200	1 in 104800
		General population	99%	1 in 248	1 in 24700	1 in 98800
EDA	Hypohidrotic ectodermal dysplasia	General population (Female)	99%	~1 in 5000	N/A	N/A
EDAR	Hypohidrotic ectodermal dysplasia 1B	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
EIF2AK3	Wolcott-Rallison syndrome	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
EIF2B5	Leukoencephalopathy with vanishing white matter	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 457	1 in 45600	1 in 182400
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
EMD	Emery-Dreifuss muscular dystrophy	General population	99%	N/A	N/A	N/A
ERCC2	Xeroderma pigmentosum	Ashkenazi Jewish	99%	1 in 107	1 in 10600	1 in 42400
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 202	1 in 20100	1 in 80400
		General population	99%	1 in 312	1 in 31100	1 in 124400
ERCC3	Xeroderma pigmentosum	Ashkenazi Jewish	99%	1 in 55	1 in 5400	1 in 21600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	1 in 388	1 in 19350	1 in 77400
		General population	98%	1 in 318	1 in 15850	1 in 63400
ERCC4	Xeroderma pigmentosum	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	1 in 441	1 in 22000	1 in 88000
ERCC5	Xeroderma pigmentosum	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ERCC6	Cockayne syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 265	1 in 26400	1 in 105600
		Non-Finnish European/White	99%	1 in 331	1 in 33000	1 in 132000
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600

ERCC8	Cockayne syndrome	Ashkenazi Jewish	99%	1 in 240	1 in 23900	1 in 95600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ESCO2	Roberts syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
ETFA	Glutaric acidemia type II	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ETFB	Glutaric acidemia type II	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ETFDH	Glutaric acidemia type II	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 411	1 in 41000	1 in 164000
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ETHE1	Ethylmalonic encephalopathy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
EVC	Ellis-van Creveld syndrome	General population	98%	1 in 142	1 in 7050	1 in 28200
EVC2	Ellis-van Creveld syndrome	Ashkenazi Jewish	99%	1 in 297	1 in 29600	1 in 118400
		Finnish	97%	1 in 318	1 in 10570	1 in 42280
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	1 in 422	1 in 42100	1 in 168400
EXOSC3	Pontocerebellar hypoplasia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 406	1 in 40500	1 in 162000
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
EYS	Retinitis pigmentosa	Ashkenazi Jewish	99%	1 in 42	1 in 4100	1 in 16400
		Finnish	98%	1 in 39	1 in 1900	1 in 7600
		Non-Finnish European/White	99%	1 in 106	1 in 10500	1 in 42000
		General population	98%	1 in 85	1 in 4200	1 in 16800
F11	Factor XI deficiency	General population	98%	1 in 500	1 in 24950	1 in 99800
		Ashkenazi Jewish	98%	1 in 11	1 in 500	1 in 2000
F2	Prothrombin Thrombophilia (F2-related)	General population	99%	1 in 150	1 in 14900	1 in 59600
F8	Hemophilia A (X-linked)	Non-Finnish European/White (Females)	80%	1 in 3 500	N/A	N/A
F9	Factor IX deficiency (hemophilia B)	General population (Female)	99%	~1 in 20000	N/A	N/A

FAH	Tyrosinemia type I	Ashkenazi Jewish	99%	1 in 137	1 in 13600	1 in 54400
		Finnish	99%	1 in 327	1 in 32600	1 in 130400
		Non-Finnish European/White	99%	1 in 309	1 in 30800	1 in 123200
		General population	99%	1 in 292	1 in 29100	1 in 116400
FAM161A	Retinitis pigmentosa	Ashkenazi Jewish	97%	1 in 236	1 in 7830	1 in 31320
		Finnish	97%	1 in 469	1 in 15600	1 in 62400
		Non-Finnish European/White	98%	1 in 356	1 in 17750	1 in 71000
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
FANCA	Fanconi anemia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 308	1 in 30700	1 in 122800
		Non-Finnish European/White	99%	1 in 174	1 in 17300	1 in 69200
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
FANCC	Fanconi anemia	Ashkenazi Jewish	99%	1 in 80	1 in 7900	1 in 31600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	1 in 470	1 in 23450	1 in 93800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
FANCG	Fanconi anemia	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
FH	Fumarase deficiency	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
		Ashkenazi Jewish	99%	~1 in 100	1 in 9900	1 in 39600
FKRP	Limb-girdle muscular dystrophy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 272	1 in 27100	1 in 108400
		Non-Finnish European/White	99%	1 in 190	1 in 18900	1 in 75600
		General population	99%	1 in 246	1 in 24500	1 in 98000
FKTN	Walker-Warburg syndrome and other FKTN- related	Ashkenazi Jewish	97%	1 in 63	1 in 2070	1 in 8280
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
G6PC	Glycogen Storage Disease Type 1A	Ashkenazi Jewish	99%	1 in 70	1 in 6900	1 in 27600
		General population	>95%	1 in 137	1 in 140	1 in 560
G6PD	Glucose-6-phosphate dehydrogenase deficiency	General population (Female)	99%	1 in 150	1 in 14900	1 in 30000 (Male)
GAA	Pompe disease	Ashkenazi Jewish	99%	1 in 77	1 in 7600	1 in 30400
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 59	1 in 5800	1 in 23200
		General population	99%	1 in 77	1 in 7600	1 in 30400
GALC	Krabbe disease	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 254	1 in 25300	1 in 101200
		General population	99%	1 in 245	1 in 24400	1 in 97600
GALE	Galactosemia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520

		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
GALK1	Galactosemia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
GALNS	Mucopolysaccharidosis type IVA	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	1 in 469	1 in 23400	1 in 93600
		Non-Finnish European/White	99%	1 in 428	1 in 42700	1 in 170800
		General population	99%	1 in 307	1 in 30600	1 in 122400
GALNT3	Hyperphosphatemic familial tumoral calcinosis	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
GALT	Galactosemia, classic	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	97%	1 in 131	1 in 4330	1 in 17320
		General population	97%	1 in 264	1 in 8770	1 in 35080
GAMT	Cerebral creatine deficiency syndromes	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
GBA	Gaucher Disease	Ashkenazi Jewish	>95%	1 in 15	1 in 20	1 in 80
		Ashkenazi Jewish	90%	1 in 15	1 in 141	1 in 560
		General population	90%	1 in 120	1 in 1200	1 in 4800
GBE1	Glycogen storage disease type IV	Ashkenazi Jewish	99%	1 in 72	1 in 7100	1 in 28400
		Finnish	99%	1 in 374	1 in 37300	1 in 149200
		Non-Finnish European/White	99%	1 in 242	1 in 24100	1 in 96400
		General population	99%	1 in 149	1 in 14800	1 in 59200
GCDH	Glutaric acidemia type I	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 356	1 in 35500	1 in 142000
		Non-Finnish European/White	99%	1 in 204	1 in 20300	1 in 81200
		General population	99%	1 in 283	1 in 28200	1 in 112800
GDF5	Du Pan syndrome	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
GFM1	Combined oxidative phosphorylation deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	1 in 432	1 in 21550	1 in 86200
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
GH1	Isolated GH deficiency, type IA	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
GHRHR	Isolated GH deficiency, type IB	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
GCH1	Dopa-responsive dystonia (AR form)	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
GJB1	Charcot-Marie-Tooth, X-linked 1	General population	90%	1 in 667	1 in 6660	1 in 26640
GJB2	Deafness and hearing loss, nonsyndromic	Ashkenazi Jewish	98%	1 in 12	1 in 550	1 in 2200

		Finnish	99%	1 in 16	1 in 1500	1 in 6000
		Non-Finnish European/White	99%	1 in 18	1 in 1700	1 in 6800
		General population	99%	1 in 20	1 in 1900	1 in 7600
GJB3	Deafness, autosomal dominant 1B	General population	99%	1 in 42	1 in 4100	1 in 16400
GJB6	Deafness, digenic (GJB2/GJB6)	General population	99%	1 in 423	1 in 42200	1 in 168800
GLA	Fabry disease	General population (Female)	99%	~1 in 5000	N/A	N/A
GLB1	GM1 gangliosidosis and mucopolysaccharidosis type IVB	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	1 in 258	1 in 12850	1 in 51400
		Non-Finnish European/White	99%	1 in 326	1 in 32500	1 in 130000
		General population	99%	1 in 337	1 in 33600	1 in 134400
GLDC	Glycine encephalopathy	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	1 in 247	1 in 24600	1 in 98400
		Non-Finnish European/White	99%	1 in 311	1 in 31000	1 in 124000
		General population	99%	1 in 372	1 in 37100	1 in 148400
GLE1	Congenital arthrogryposis with anterior horn cell disease	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 42	1 in 4100	1 in 16400
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	1 in 442	1 in 44100	1 in 176400
GNE	Inclusion body myopathy 2	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 374	1 in 37300	1 in 149200
		General population	99%	1 in 220	1 in 21900	1 in 87600
GNPTAB	Mucolipidosis type II and III	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	1 in 165	1 in 5470	1 in 21880
		Non-Finnish European/White	98%	1 in 258	1 in 12850	1 in 51400
		General population	97%	1 in 295	1 in 9800	1 in 39200
GNPTG	Mucolipidosis III gamma	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	97%	< 1 in 500	<1 in 16630	<1 in 66520
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
GNS	Mucopolysaccharidosis type III	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
GORAB	Geroderma osteodysplastica	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
GP1BA	Bernard-Soulier syndrome, type A1	General population	98%	1 in 500	1 in 24950	1 in 99800
GP1BB	Bernard-Soulier syndrome, type B	General population	98%	1 in 500	1 in 24950	1 in 99800
GP9	Bernard-Soulier syndrome, type C	General population	98%	1 in 500	1 in 24950	1 in 99800
GRHPR	Primary hyperoxaluria	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	97%	1 in 469	1 in 15600	1 in 62400
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
GUCY2D	Leber congenital amaurosis 1	General population	98%	<1 in 500	<1 in 24950	<1 in 99800

GUSB	Mucopolysaccharidosis type VII	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 127	1 in 12600	1 in 50400
		Non-Finnish European/White	99%	1 in 216	1 in 21500	1 in 86000
		General population	99%	1 in 249	1 in 24800	1 in 99200
HADHB	Mitochondrial trifunctional protein deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
HAX1	Severe congenital neutropenia	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	97%	< 1 in 500	<1 in 16630	<1 in 66520
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
HBA1/HBA2	Alpha-thalassemia	Non-Finnish European/White	97%	1 in 20	1 in 630	1 in 2500
HBB	Beta-hemoglobinopathies, includes sickle cell disease	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	99%	1 in 256	1 in 25500	1 in 102000
		General population	99%	1 in 116	1 in 11500	1 in 46000
HEXA	Tay-Sachs disease	Ashkenazi Jewish	97%	1 in 31	1 in 1000	1 in 4000
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	99%	1 in 265	1 in 26400	1 in 105600
		General population	97%	1 in 252	1 in 8370	1 in 33480
HEXB	Sandhoff disease	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 278	1 in 27700	1 in 110800
		General population	98%	1 in 467	1 in 23300	1 in 93200
HFE	Hemochromatosis (type I)	General population	100%	1 in 32	N/A	
HGD	Alkaptonuria	General population	90%	1 in 250	1 in 2490	1 in 9960
HGSNAT	Mucopolysaccharidosis type III	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 482	1 in 48100	1 in 192400
		General population	99%	1 in 485	1 in 48400	1 in 193600
HJV	Juvenile hereditary hemochromatosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
HLCS	Holocarboxylase synthetase deficiency	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600

		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
HMGCL	HMG-CoA lyase deficiency	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
HMOX1	Heme oxygenase-1 deficiency	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
HOGA1	Primary hyperoxaluria	Ashkenazi Jewish	97%	1 in 48	1 in 1570	1 in 6280
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 183	1 in 18200	1 in 72800
		General population	99%	1 in 136	1 in 13500	1 in 54000
HPD	Tyrosinemia type III	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
HPS1	Hermansky-Pudlak syndrome	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
HPS3	Hermansky-Pudlak syndrome	Ashkenazi Jewish	99%	1 in 287	1 in 28600	1 in 114400
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	1 in 489	1 in 24400	1 in 97600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
HPS4	Hermansky-Pudlak syndrome	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
HSD17B3	17-HSD3 deficiency (46,XY DSD)	Non-Finnish European/White	99%	1 in 200	1 in 20000	1 in 80000
HSD17B4	D-bifunctional protein deficiency	Non-Finnish European/White	97%	1 in 300	1 in 10000	1 in 40000
HSD3B2	Congenital adrenal hyperplasia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
HYLS1	Hydrolethalus syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
CHM	Choroideremia	General population	95%	<1 in 500	<1 in 9980	<1 in 39920
CHRNE	Congenital myasthenic syndrome	Ashkenazi Jewish	97%	1 in 153	1 in 5070	1 in 20280
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
CHRNG	Multiple pterygium syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 466	1 in 46500	1 in 186000
		Non-Finnish European/White	97%	1 in 338	1 in 11230	1 in 44920

		General population	97%	1 in 452	1 in 15030	1 in 60120
IDS	Mucopolysaccharidosis type II	General population	99%	N/A	N/A	N/A
IDUA	Mucopolysaccharidosis type I	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	1 in 178	1 in 17700	1 in 70800
		Non-Finnish European/White	99%	1 in 98	1 in 9700	1 in 38800
		General population	99%	1 in 354	1 in 35300	1 in 141200
IL2RG	Severe combined Immunodeficiency (SCID), X-linked	General population (Female)	99%	~1 in 25000	N/A	N/A
ITGB3	Glanzmann thrombasthenia	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
IVD	Isovaleric acidemia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 273	1 in 27200	1 in 108800
		General population	99%	1 in 349	1 in 34800	1 in 139200
KCNJ11	Familial hyperinsulinism	General population	99%	1 in 423	1 in 42200	1 in 168800
LAMA2	Muscular dystrophy, LAMA2-related	Ashkenazi Jewish	99%	1 in 296	1 in 29500	1 in 118000
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	1 in 206	1 in 10250	1 in 41000
		General population	99%	1 in 414	1 in 41300	1 in 165200
LAMA3	Junctional epidermolysis bullosa	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
LAMB3	Junctional epidermolysis bullosa	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 243	1 in 24200	1 in 96800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
LAMC2	Junctional epidermolysis bullosa	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
LCA5	Leber congenital amaurosis	Ashkenazi Jewish	99%	1 in 219	1 in 21800	1 in 87200
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
LDLR	Familial hypercholesterolemia	General population	99%	1 in 200	1 in 19900	1 in 79600
		Ashkenazi Jewish	99%	1 in 70	1 in 6900	1 in 27600
LDLRAP1	Familial hypercholesterolemia (AR form)	General population	99%	1 in 8	1 in 700	1 in 2800
LHCGR	Leydig cell hypoplasia	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
LIFR	Stüve-Wiedemann syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520

LIPA	Lysosomal acid lipase deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 232	1 in 23100	1 in 92400
		General population	99%	1 in 343	1 in 34200	1 in 136800
LIPH	Woolly hair/hypotrichosis	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
LOXHD1	Deafness and hearing loss, nonsyndromic	Ashkenazi Jewish	99%	1 in 130	1 in 12900	1 in 51600
		Finnish	99%	1 in 301	1 in 30000	1 in 120000
		Non-Finnish European/White	99%	1 in 152	1 in 15100	1 in 60400
		General population	99%	1 in 414	1 in 41300	1 in 165200
LPL	Lipoprotein lipase deficiency, familial	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
LRPPRC	Leigh syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	98%	1 in 326	1 in 16250	1 in 65000
LYST	Chediak-Higashi syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
MAN2B1	Alpha-mannosidosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 206	1 in 20500	1 in 82000
		Non-Finnish European/White	99%	1 in 426	1 in 42500	1 in 170000
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
MAT1A	Hypermethioninemia	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
MCCC1	3-Methylcrotonyl-CoA carboxylase deficiency	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 444	1 in 44300	1 in 177200
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency	Ashkenazi Jewish	99%	1 in 192	1 in 19100	1 in 76400
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 249	1 in 24800	1 in 99200
		General population	99%	1 in 168	1 in 16700	1 in 66800
MCOLN1	Mucolipidosis type IV	Ashkenazi Jewish	99%	1 in 118	1 in 11700	1 in 46800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
MECP2	Rett syndrome (XL)	General population	99%			
		General population	99%	N/A	N/A	N/A
MED17	Microcephaly, postnatal progressive, with seizures	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600

		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
MEFV	Familial Mediterranean fever	Ashkenazi Jewish	99%	1 in 10	1 in 900	1 in 3600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 127	1 in 12600	1 in 50400
		General population	99%	1 in 56	1 in 5500	1 in 22000
MESP2	Spondylothoracic dysostosis	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	1 in 124	1 in 4100	1 in 16400
		Non-Finnish European/White	97%	1 in 233	1 in 7730	1 in 30920
		General population	97%	1 in 48	1 in 1570	1 in 6280
MFSD8	Neuronal ceroid-lipofuscinosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
MKKS	Bardet-Biedl syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
MKS1	Ciliopathies	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	1 in 72	1 in 2370	1 in 9480
		Non-Finnish European/White	98%	1 in 266	1 in 13250	1 in 53000
		General population	97%	1 in 232	1 in 7700	1 in 30800
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts type 1	Ashkenazi Jewish	99%	1 in 192	1 in 19100	1 in 76400
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
MLYCD	Malonyl-CoA decarboxylase deficiency	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
MMAA	Methylmalonic acidemia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
MMAB	Methylmalonic acidemia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	98%	1 in 442	1 in 22050	1 in 88200
MMADHC	Methylmalonic acidemia with homocystinuria	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
MMACHC	Methylmalonic acidemia with homocystinuria	Ashkenazi Jewish	97%	1 in 179	1 in 5930	1 in 23720
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	1 in 167	1 in 8300	1 in 33200
		General population	98%	1 in 162	1 in 8050	1 in 32200

MMUT	Methylmalonic acidemia	Ashkenazi Jewish	99%	1 in 316	1 in 31500	1 in 126000
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 329	1 in 32800	1 in 131200
		General population	99%	1 in 365	1 in 36400	1 in 145600
MOCS1	Molybdenum cofactor deficiency	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
MPI	Congenital disorders of glycosylation	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	1 in 323	1 in 32200	1 in 128800
MPL	Congenital amegakaryocytic thrombocytopenia	Ashkenazi Jewish	99%	1 in 59	1 in 5800	1 in 23200
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 278	1 in 27700	1 in 110800
		General population	99%	1 in 301	1 in 30000	1 in 120000
MPV17	Mitochondrial DNA depletion syndrome, MPV17-related	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
MRE11	Ataxia-telangiectasia-like disorder 1	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
MTHFR	Homocystinuria due to MTHFR def.	General population	98%	1 in 224	1 in 11150	1 in 44600
MTM1	Myotubular myopathy	General population (Female)	99%	~1 in 25000	N/A	N/A
MTRR	Homocystinuria	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	97%	1 in 467	1 in 15530	1 in 62120
MTTP	Abetalipoproteinemia	Ashkenazi Jewish	99%	1 in 187	1 in 18600	1 in 74400
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
MYO15A	Deafness, autosomal recessive 3	General population	98%	1 in 500	1 in 24950	1 in 99800
MYO7A	Usher syndrome (hearing loss and retinitis pigmentosa	Ashkenazi Jewish	99%	1 in 380	1 in 37900	1 in 151600
		Finnish	97%	1 in 319	1 in 10600	1 in 42400
		Non-Finnish European/White	99%	1 in 158	1 in 15700	1 in 62800
		General population	98%	1 in 270	1 in 13450	1 in 53800
NAGLU	Mucopolysaccharidosis type III	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 344	1 in 34300	1 in 137200
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600

NAGS	N-acetylglutamate synthetase deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
NBN	Nijmegen breakage syndrome	Ashkenazi Jewish	99%	1 in 435	1 in 43400	1 in 173600
		Finnish	99%	1 in 399	1 in 39800	1 in 159200
		Non-Finnish European/White	97%	< 1 in 500	<1 in 16630	<1 in 66520
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
NDRG1	Charcot-Marie-Tooth disease 4D	General population	98%	1 in 22	1 in 1050	1 in 4200
NDUFAF5	Leigh syndrome	Ashkenazi Jewish	99%	1 in 143	1 in 14200	1 in 56800
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 383	1 in 38200	1 in 152800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
NDUFS4	Leigh syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	97%	< 1 in 500	<1 in 16630	<1 in 66520
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
NDUFS6	Leigh syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
NEB	Nemaline myopathy	Ashkenazi Jewish	99%	1 in 304	1 in 30300	1 in 121200
		Finnish	99%	1 in 139	1 in 13800	1 in 55200
		Non-Finnish European/White	92%	1 in 137	1 in 1700	1 in 6800
		General population	99%	1 in 133	1 in 13200	1 in 52800
NEU1	Sialidosis	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
NLRP7	Hydatidiform mole, recurrent	General population	95%	<1 in 500	<1 in 9980	<1 in 39920
NPC1	Niemann-Pick disease type C	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	98%	1 in 438	1 in 21850	1 in 87400
		Non-Finnish European/White	99%	1 in 231	1 in 23000	1 in 92000
		General population	99%	1 in 359	1 in 35800	1 in 143200
NPC2	Niemann-Pick disease type C	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
NPHP1	Joubert syndrome and related disorders	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
NPHS1	Nephrotic syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520

		Finnish	97%	1 in 38	1 in 1230	1 in 4920
		Non-Finnish European/White	99%	1 in 206	1 in 20500	1 in 82000
		General population	99%	1 in 213	1 in 21200	1 in 84800
NPHS2	Nephrotic syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 261	1 in 26000	1 in 104000
		General population	99%	1 in 386	1 in 38500	1 in 154000
NR0B1	Congenital adrenal hypoplasia, X-linked	General population	99%	N/A	N/A	N/A
NR2E3	Enhanced S-cone syndrome	General population	98%	1 in 209	1 in 10400	1 in 41600
NTRK1	Congenital insensitivity to pain with anhidrosis	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
OAT	Gyrate atrophy of choroid and retina	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
OCRL	Dent disease	General population	99%	N/A	N/A	N/A
OPA3	Costeff syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
OTC	Ornithine transcarbamylase deficiency	General population (Female)	99%	~1 in 20000	N/A	N/A
PAH	Phenylalanine hydroxylase deficiency	Ashkenazi Jewish	99%	1 in 17	1 in 1600	1 in 6400
		Finnish	99%	1 in 172	1 in 17100	1 in 68400
		Non-Finnish European/White	99%	1 in 39	1 in 3800	1 in 15200
		General population	99%	1 in 41	1 in 4000	1 in 16000
PANK2	Pantothenate kinase-associated neurodegeneration	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PC	Pyruvate carboxylase deficiency	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PCCA	Propionic acidemia	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
PCCB	Propionic acidemia	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800

		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PCDH15	Usher syndrome (hearing loss and retinitis pigmentosa	Ashkenazi Jewish	99%	1 in 116	1 in 11500	1 in 46000
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	1 in 397	1 in 19800	1 in 79200
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
PDHA1	Pyruvate dehydrogenase deficiency	General population (Female)	99%	~1 in 10000	N/A	N/A
PDHB	Pyruvate dehydrogenase deficiency	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PEPD	Prolidase deficiency	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
PET100	Mitochondrial complex IV deficiency	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
PEX1	Zellweger spectrum disorder/ peroxisome biogenesis disorder	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	1 in 204	1 in 10150	1 in 40600
		General population	98%	1 in 393	1 in 19600	1 in 78400
PEX10	Zellweger spectrum disorder/ peroxisome biogenesis disorder	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PEX12	Zellweger spectrum disorder/ peroxisome biogenesis disorder	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
PEX2	Zellweger spectrum disorder/ peroxisome biogenesis disorder	Ashkenazi Jewish	99%	1 in 208	1 in 20700	1 in 82800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PEX6	Zellweger spectrum disorder/ peroxisome biogenesis disorder	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	1 in 470	1 in 23450	1 in 93800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PEX7	Rhizomelic chondrodysplasia punctata	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 429	1 in 42800	1 in 171200
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PFKM	Glycogen storage disease type VII	Ashkenazi Jewish	98%	1 in 100	1 in 4950	1 in 19800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800

		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PHGDH	Phosphoglycerate dehydrogenase deficiency	Ashkenazi Jewish	99%	1 in 324	1 in 32300	1 in 129200
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PIGN	MCAHS1	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
PKHD1	Polycystic kidney disease, autosomal recessive	Ashkenazi Jewish	97%	1 in 62	1 in 2030	1 in 8120
		Finnish	99%	1 in 38	1 in 3700	1 in 14800
		Non-Finnish European/White	99%	1 in 92	1 in 9100	1 in 36400
		General population	99%	1 in 96	1 in 9500	1 in 38000
PLA2G6	Neurodegeneration with brain iron accumulation disorder	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 474	1 in 47300	1 in 189200
		General population	98%	1 in 403	1 in 20100	1 in 80400
PMM2	Congenital disorders of glycosylation	Ashkenazi Jewish	99%	1 in 65	1 in 6400	1 in 25600
		Finnish	99%	1 in 58	1 in 5700	1 in 22800
		Non-Finnish European/White	99%	1 in 63	1 in 6200	1 in 24800
		General population	99%	1 in 81	1 in 8000	1 in 32000
PNPO	Pyridoxal 5'-phosphate-dependent epilepsy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
POLG	POLG-related disorders	Ashkenazi Jewish	99%	1 in 115	1 in 11400	1 in 45600
		Finnish	99%	1 in 62	1 in 6100	1 in 24400
		Non-Finnish European/White	99%	1 in 49	1 in 4800	1 in 19200
		General population	99%	1 in 67	1 in 6600	1 in 26400
POLH	Xeroderma pigmentosum	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
POMGNT1	Limb-girdle muscular dystrophy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 202	1 in 20100	1 in 80400
		Non-Finnish European/White	99%	1 in 313	1 in 31200	1 in 124800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
POR	Congenital adrenal hyperplasia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	1 in 273	1 in 13600	1 in 54400
		Non-Finnish European/White	99%	1 in 312	1 in 31100	1 in 124400
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PPT1	Neuronal ceroid-lipofuscinosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 74	1 in 7300	1 in 29200
		Non-Finnish European/White	99%	1 in 304	1 in 30300	1 in 121200

		General population	99%	1 in 190	1 in 18900	1 in 75600
PREPL	Hypotonia-cystinuria syndrome	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
PROP1	Combined pituitary hormone deficiency	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PRPS1	Arts syndrome (XL)	General population	98%	N/A	N/A	N/A
PSAP	Metachromatic leukodystrophy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PTS	Tetrahydrobiopterin deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PUS1	Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
PYGM	Glycogen storage disease type V	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 496	1 in 49500	1 in 198000
		Non-Finnish European/White	99%	1 in 128	1 in 12700	1 in 50800
		General population	99%	1 in 167	1 in 16600	1 in 66400
RAB23	Carpenter syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
RAG1	Omenn syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 421	1 in 42000	1 in 168000
		General population	99%	1 in 331	1 in 33000	1 in 132000
RAG2	Omenn syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
RAPSN	Congenital myasthenic syndrome	Ashkenazi Jewish	99%	1 in 247	1 in 24600	1 in 98400
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 169	1 in 16800	1 in 67200
		General population	99%	1 in 451	1 in 45000	1 in 180000
RARS2	Pontocerebellar hypoplasia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	1 in 364	1 in 18150	1 in 72600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
RDH12	Leber congenital amaurosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600

		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	1 in 432	1 in 43100	1 in 172400
RLBP1	Retinitis pigmentosa	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
RMRP	Cartilage-hair hypoplasia	Ashkenazi Jewish	99%	1 in 70	1 in 6900	1 in 27600
		Finnish	99%	1 in 55	1 in 5400	1 in 21600
		Non-Finnish European/White	99%	1 in 292	1 in 29100	1 in 116400
		General population	98%	1 in 92	1 in 4550	1 in 18200
RNASEH2C	Aicardi-Goutières syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
RPE65	Leber congenital amaurosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 412	1 in 41100	1 in 164400
		General population	99%	1 in 413	1 in 41200	1 in 164800
RPGRIP1L	Joubert syndrome and related disorders, including Meckel-Gruber syndrome	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 301	1 in 30000	1 in 120000
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
RS1	Juvenile retinoschisis, X-linked	General population (Female)	99%	~1 in 12500	N/A	N/A
RTEL1	Dyskeratosis congenita 5	General population	99%	1 in 500	1 in 49900	1 in 199600
		Ashkenazi Jewish	99%	1 in 200	1 in 19900	1 in 79600
SACS	Autosomal recessive spastic ataxia of Charlevoix	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	1 in 307	1 in 15300	1 in 61200
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SAMD9	MIRAGE syndrome	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
SAMHD1	Aicardi-Goutières syndrome	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SBDS	Shwachman-Diamond syndrome	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
SEPSECS	Pontocerebellar hypoplasia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 163	1 in 16200	1 in 64800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SERPINA1	Alpha-1 antitrypsin deficiency	Non-Finnish European/White	97%	1 in 25	1 in 800	1 in 3200
SGCA	Limb-girdle muscular dystrophy	Ashkenazi Jewish	99%	1 in 283	1 in 28200	1 in 112800
		Finnish	99%	1 in 266	1 in 26500	1 in 106000
		Non-Finnish European/White	99%	1 in 348	1 in 34700	1 in 138800

		General population	99%	1 in 341	1 in 34000	1 in 136000
SGCB	Limb-girdle muscular dystrophy	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SGCD	Limb-girdle muscular dystrophy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SGCG	Limb-girdle muscular dystrophy	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SGSH	Mucopolysaccharidosis type III	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 463	1 in 46200	1 in 184800
		Non-Finnish European/White	99%	1 in 232	1 in 23100	1 in 92400
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SLC12A3	Gitelman syndrome	Non-Finnish European/White	99%	1 in 80	1 in 7900	1 in 31600
SLC12A6	Andermann syndrome (ACCPN)	Non-Finnish European/White	99%	<1 in 500	<1 in 49900	<1 in 199600
SLC17A5	Sialic acid storage disorders	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 83	1 in 8200	1 in 32800
		Non-Finnish European/White	99%	1 in 362	1 in 36100	1 in 144400
		General population	98%	1 in 479	1 in 23900	1 in 95600
SLC19A2	Thiamine-responsive megaloblastic anemia	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
SLC22A5	Systemic primary carnitine deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 227	1 in 22600	1 in 90400
		Non-Finnish European/White	99%	1 in 114	1 in 11300	1 in 45200
		General population	99%	1 in 211	1 in 21000	1 in 84000
SLC25A13	Citrin deficiency (CTLN2)	Non-Finnish European/White	97%	<1 in 500	<1 in 49900	<1 in 199600
SLC25A15	HHH syndrome	Non-Finnish European/White	99%	<1 in 500	<1 in 49900	<1 in 199600
SLC25A20	Carnitine-Acylcarnitine Translocase Deficiency (CACT)	Non-Finnish European/White	99%	1 in 500	1 in 49901	1 in 199904
SLC26A2	Diastrophic dysplasia / atelosteogenesis	Non-Finnish European/White	97%	1 in 200	1 in 6600	1 in 26500
		Finnish	97%	1 in 70	1 in 2300	1 in 9200
SLC26A3	Congenital chloride diarrhea	Non-Finnish European/White	99%	<1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 50	1 in 4900	1 in 19600
SLC26A4	Pendred syndrome / DFNB4 hearing loss	Non-Finnish European/White	97%	1 in 80	1 in 2600	1 in 10500
SLC35A3	Arthrogryposis, mental retardation, and seizures	Ashkenazi Jewish	99%	1 in 373	1 in 37200	1 in 148800
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600

SLC37A4	Glycogen storage disease type I	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	1 in 481	1 in 24000	1 in 96000
SLC39A4	Acrodermatitis enteropathica	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	1 in 278	1 in 9230	1 in 36920
		Non-Finnish European/White	98%	1 in 386	1 in 19250	1 in 77000
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
SLC3A1	Cystinuria type A	General population	98%	1 in 50	1 in 2450	1 in 9800
SLC45A2	Oculocutaneous albinism 4	General population	98%	1 in 159	1 in 7900	1 in 31600
SLC4A11	Corneal dystrophy and perceptive deafness	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
SLC6A8	Cerebral creatine deficiency syndromes	General population (Female)	99%	~1 in 5000	N/A	N/A
SLC7A7	Lysinuric protein intolerance	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	1 in 110	1 in 10900	1 in 43600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SLC7A9	Cystinuria type B	General population	98%	1 in 42	1 in 2050	1 in 8200
SMARCAL1	Schimke immunoosseous dysplasia	Ashkenazi Jewish	99%	1 in 192	1 in 19100	1 in 76400
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SMN1	Spinal muscular atrophy	General population	91%	1 in 54	1 in 590	1 in 2360
SMPD1	Niemann-Pick disease types A and B	Ashkenazi Jewish	99%	1 in 107	1 in 10600	1 in 42400
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	99%	1 in 460	1 in 45900	1 in 183600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SRD5A2	5-alpha-reductase deficiency	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
ST3GAL5	Nanism with neurological involvement	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
STAR	Lipoid congenital adrenal hyperplasia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
STRC	Deafness, autosomal recessive 16	General population	95%	1 in 200	1 in 3980	1 in 15920
SUCLA2	Mitochondrial DNA depletion syndrome 5	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
SUMF1	Multiple sulfatase deficiency	Ashkenazi Jewish	99%	1 in 273	1 in 27200	1 in 108800

		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
SURF1	Leigh syndrome	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	1 in 341	1 in 17000	1 in 68000
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
TAT	Tyrosinemia type II	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
TCIRG1	Osteopetrosis	Ashkenazi Jewish	99%	1 in 417	1 in 41600	1 in 166400
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 417	1 in 41600	1 in 166400
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
TECPR2	Hereditary spastic paraplegia	Ashkenazi Jewish	97%	1 in 155	1 in 5130	1 in 20520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
TFR2	Hereditary hemochromatosis	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
TGM1	Autosomal recessive congenital ichthyosis (ARCI)	Ashkenazi Jewish	99%	1 in 463	1 in 46200	1 in 184800
		Finnish	99%	1 in 195	1 in 19400	1 in 77600
		Non-Finnish European/White	99%	1 in 221	1 in 22000	1 in 88000
		General population	99%	1 in 265	1 in 26400	1 in 105600
TH	Tyrosine hydroxylase deficiency	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
TMC1	Deafness, autosomal recessive 7	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
TMEM216	Joubert syndrome and related disorders, including Meckel-Gruber syndrome	Ashkenazi Jewish	99%	1 in 148	1 in 14700	1 in 58800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
TPO	Congenital hypothyroidism 2	General population	99%	1 in 373	1 in 37200	1 in 148800
TPP1	Neuronal ceroid-lipofuscinosis	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	1 in 281	1 in 14000	1 in 56000
		Non-Finnish European/White	99%	1 in 281	1 in 28000	1 in 112000
		General population	99%	1 in 318	1 in 31700	1 in 126800
TREX1	Aicardi-Goutières syndrome 1	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
TRIM32	Limb-girdle muscular dystrophy	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	97%	< 1 in 500	<1 in 16630	<1 in 66520

		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
TRIM37	Mulibrey nanism	Finnish	99%	<1 in 500	<1 in 49900	<1 in 199600
		General population	99%	1 in 26	1 in 2500	1 in 10000
TRMU	Acute infantile liver failure	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
TSEN54	Pontocerebellar hypoplasia	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	1 in 242	1 in 24100	1 in 96400
		General population	99%	1 in 306	1 in 30500	1 in 122000
TSFM	Combined oxidative phosphorylation deficiency	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	1 in 35	1 in 3400	1 in 13600
		Non-Finnish European/White	99%	1 in 483	1 in 48200	1 in 192800
		General population	99%	1 in 307	1 in 30600	1 in 122400
TSHB	Central congenital hypothyroidism 1	General population	99%	1 in 500	1 in 49900	1 in 199600
TSHR	Congenital hypothyroidism 1	General population	99%	1 in 500	1 in 49900	1 in 199600
TTC37	Trichohepatoenteric syndrome	General population	98%	1 in 500	1 in 24950	1 in 99800
TTN	Dilated cardiomyopathy 1G (AR form)	General population	95%	<1 in 500	<1 in 9980	<1 in 39920
TTPA	Ataxia with vitamin E deficiency	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
TYMP	Mitochondrial neurogastrointestinal encephalopathy	Ashkenazi Jewish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	1 in 410	1 in 40900	1 in 163600
TYR	Oculocutaneous albinism	Ashkenazi Jewish	99%	1 in 21	1 in 2000	1 in 8000
		Finnish	99%	1 in 42	1 in 4100	1 in 16400
		Non-Finnish European/White	99%	1 in 51	1 in 5000	1 in 20000
		General population	99%	1 in 49	1 in 4800	1 in 19200
TYRP1	Oculocutaneous albinism 3	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
UGT1A1	Crigler-Najjar syndrome	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
UPB1	Beta-ureidopropionase deficiency	General population	98%	<1 in 500	<1 in 24950	<1 in 99800
USH1C	Usher syndrome (hearing loss and retinitis pigmentosa	Ashkenazi Jewish	97%	1 in 235	1 in 7800	1 in 31200
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
USH2A	Usher syndrome (hearing loss and retinitis pigmentosa	Ashkenazi Jewish	99%	1 in 44	1 in 4300	1 in 17200
		Finnish	98%	1 in 142	1 in 7050	1 in 28200
		Non-Finnish European/White	99%	1 in 55	1 in 5400	1 in 21600

		General population	99%	1 in 61	1 in 6000	1 in 24000
VPS13A	Choreacanthocytosis	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	1 in 352	1 in 17550	1 in 70200
		General population	98%	1 in 420	1 in 20950	1 in 83800
VPS13B	Cohen syndrome	Ashkenazi Jewish	97%	1 in 280	1 in 9300	1 in 37200
		Finnish	97%	1 in 123	1 in 4070	1 in 16280
		Non-Finnish European/White	98%	1 in 225	1 in 11200	1 in 44800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
VPS45	Severe congenital neutropenia	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	99%	< 1 in 500	<1 in 49900	<1 in 199600
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
VPS53	Pontocerebellar hypoplasia	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	97%	< 1 in 500	<1 in 16630	<1 in 66520
VRK1	Pontocerebellar hypoplasia	Ashkenazi Jewish	99%	1 in 297	1 in 29600	1 in 118400
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	1 in 202	1 in 20100	1 in 80400
VSX2	Microphthalmia 2	General population	98%	1 in 91	1 in 4500	1 in 18000
VWF	Von Willebrand disease 3	General population	70%	<1 in 500	<1 in 1660	<1 in 6640
WAS	Wiskott-Aldrich syndrome	General population	99%	N/A	N/A	N/A
WNT10A	Odonto-onycho-dermal dysplasia	General population	99%	<1 in 500	<1 in 49900	<1 in 199600
WRN	Werner syndrome	Ashkenazi Jewish	99%	1 in 428	1 in 42700	1 in 170800
		Finnish	99%	< 1 in 500	<1 in 49900	<1 in 199600
		Non-Finnish European/White	98%	1 in 335	1 in 16700	1 in 66800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
XPA	Xeroderma pigmentosum	Ashkenazi Jewish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Finnish	99%	1 in 155	1 in 15400	1 in 61600
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	98%	< 1 in 500	<1 in 24950	<1 in 99800
XPC	Xeroderma pigmentosum	Ashkenazi Jewish	97%	< 1 in 500	<1 in 16630	<1 in 66520
		Finnish	98%	< 1 in 500	<1 in 24950	<1 in 99800
		Non-Finnish European/White	98%	< 1 in 500	<1 in 24950	<1 in 99800
		General population	99%	< 1 in 500	<1 in 49900	<1 in 199600
ZFYVE26	Spastic paraplegia 33	General population	98%	<1 in 500	<1 in 24950	<1 in 99800