Estimated residual risks after tested negative using Blueprint Genetics Carrier Test

Blueprint Genetics

This table below displays residual risks after a negative result for each of the genes and corresponding disorders and is not calculated separately for each tested individual. If a patient is reported to be a carrier of a disease, their residual risk equals "1" and this table does not apply for that disease.

Residual risk values were calculated based on published mutation or carrier frequencies for dominant and recessive disorders (e.g., Guo MH and Gregg AR, 2019) and estimated detection rates of Blueprint Genetics' testing technologies. Detection rate is probability to detect disease-causing variants when accounting for sequence coverage, mapping quality and known mutation profile.

Key: **AD** = autosomal dominant, **AR** = autosomal recessive; **XL** = X-linked; **XLD** = X-linked dominant, **XLR** = X-linked recessive, **Worldwide** = global, non-specific populations

Gene	Phenotypes	Inheritance model	Ethnicity	Carrier frequency	Detection rate	Residual risk	Comprehensive Reproductive Screen (FMR1 / DUO) 461 genes	Core Reproductive Screen (FMR1 / DUO) 126 genes	Ashkenazi Jewish Reproductive Screen (FMR1 / DUO) 67 genes	Comprehensive Reproductive Male Screen 433 genes	Core Reproductive Male Screen 114 genes
AAAS	Achalasia-addisonianism-alacrimia syndrome	AR	Worldwide	1 in 416	99	1 in 41580	•		•	•	
AARS2	Progressive leukoencephalopathy with ovarian failure	AR	Worldwide	1 in 514	99	1 in 51390	•	•		•	•
ABCA3	Pulmonary surfactant metabolism dysfunction type 3	AR	Worldwide	1 in 270	99	1 in 27000	•	•		•	•
ABCB11	Progressive familial intrahepatic cholestasis type 2	AR	Worldwide	1 in 200	99	1 in 20000	•			•	
ABCC6#	Pseudoxanthoma elasticum	AR	Worldwide	1 in 90	98	1 in 4500	•	•		•	•
ABCC8	Hyperinsulinemic hypoglycemia	AD/AR	Worldwide	1 in 180	99	1 in 18000	•	•	•	•	•
ABCD1#	Adrenoleukodystrophy	XLR	Worldwide	1 in 8500	98	1 in 425000	•	•			
ACAD9	Acyl-CoA dehydrogenase family, deficiency	AR	Worldwide	1 in 600	99	1 in 60000	•			•	
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency	AR	Worldwide	1 in 110	99	1 in 11000	•	•		•	•
ACADS	Short-Chain Acyl-CoA Dehydrogenase Deficiency	AR	Worldwide	1 in 127	99	1 in 12690	•			•	
ACADSB	2-Methylbutyrylglycinuria	AR	Worldwide	1 in 199	99	1 in 19890	•			•	
ACADVL	Very long chain Acyl-CoA dehydroge-nase deficiency	AR	Worldwide	1 in 200	99	1 in 20000	•	•		•	•
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	AR	Worldwide	1 in 3254	99	1 in 325400	•			•	

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ACSF3	Combined malonic and methylmalonic aciduria	AR	Worldwide	1 in 132	99	1 in 13200	•			•	
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency	AR	Worldwide	1 in 420	99	1 in 42000	•			•	
ADA2	ADA2 deficiency	AR	Worldwide	1 in 335	99	1 in 33480	•			•	
ADAMTS2 Excluded from the analysis: NM_021599, exon 11	Ehlers-Danlos syndrome	AR	Worldwide	1 in 2200	98	1 in 110000	•		•	•	
ADAMTSL4	Isolated ectopia lentis	AR	Worldwide	1 in 50	99	1 in 4950	•			•	
ADGRG1	Bilateral polymicrogyria	AR	Worldwide	1 in 980	99	1 in 98000	•			•	
ADGRV1	Usher syndrome type IIC	AR	Worldwide	1 in 192	99	1 in 19170	•	•		•	•
AGA	Aspartylglucosaminuria	AR	Worldwide	1 in 970	99	1 in 97000	•	•		•	•
AGL	Glycogen storage disease	AR	Worldwide	1 in 620	99	1 in 62000	•		•	•	
AGPS	Rhizomelic chondrodysplasia punctata type 3	AR	Worldwide	1 in 158	99	1 in 15800	•			•	
AGXT	Primary hyperoxaluria type 1	AR	Worldwide	1 in 316	99	1 in 31600	•	•		•	•
AHI1	Joubert syndrome	AR	Worldwide	1 in 2300	99	1 in 230000	•	•		•	•
AIFM1	Severe X-linked mitochondrial encephalomyopathy, Cowchock syndrome and Deafness	XLR	Worldwide	1 in 450	99	1 in 45000	•				
AIRE	Autoimmune polyendocrinopathy syndrome	AR	Worldwide	1 in 210	99	1 in 21000	•	•		•	•
ALDH3A2	Sjogren-Larsson syndrome	AR	Worldwide	1 in 1200	99	1 in 120000	•			•	
ALDH7A1	Pyridoxine Dependent Epilepsy	AR	Worldwide	1 in 294	99	1 in 29430	•			•	
ALDOB	Fructose intolerance, hereditary	AR	Worldwide	1 in 160	99	1 in 16000	•	•		•	•
ALG6	Congenital disorder of glycosylation	AR	Worldwide	1 in 920	99	1 in 92000	•			•	
ALMS1#	Alström syndrome	AR	Worldwide	1 in 1920	98	1 in 96000	•	•		•	•
ALPL	Hypophosphatasia perinatal lethal, infantile, juvenile and adult forms	AD/AR	Worldwide	1 in 588	99	1 in 58800	•	•		•	•
AMT	Glycine encephalopathy	AR	Worldwide	1 in 1240	99	1 in 124000	•			•	
ANO10	Spinocerebellar ataxia	AR	Worldwide	1 in 520	99	1 in 52000	•	•		•	•

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AP1S1	MEDNIK syndrome	AR	Worldwide	1 in 1423	99	1 in 142290	•			•	
AP3B1	Hermansky-Pudlak syndrome	AR	Worldwide	1 in 212	95	1 in 4248	•			•	
AR	Androgen insensitivity and Hypospadias 1, X-linked	XLR	Worldwide	1 in 18000	97	1 in 600000	•				
ARG1	Hyperargininemia	AR	Worldwide	1 in 296	99	1 in 29600	•			•	
ARL6	Bardet-Biedl syndrome and Retinitis pigmentosa	AR	Worldwide	1 in 712	99	1 in 71190	•			•	
ARSA	Metachromatic leukodystrophy	AR	Worldwide	1 in 288	99	1 in 28800	•	•	•	•	•
ARSB	Mucopolysaccharidosis (Maro-teaux-Lamy)	AR	Worldwide	1 in 613	99	1 in 61300	•			•	
ARX	Epileptic encephalopathy, Hydranencephaly with abnormal genitalia and Lissencephaly	XL	Worldwide	1 in 450000	90	1 in 4500000	•	•			
ASL	Argininosuccinic aciduria	AR	Worldwide	1 in 223	99	1 in 22300	•	•		•	•
ASNS#	Asparagine synthetase deficiency	AR	Worldwide	1 in 500	97	1 in 16667	•			•	
ASPA	Aspartoacylase deficiency (Canavan disease)	AR	Worldwide	1 in 427	99	1 in 42700	•	•	•	•	•
ASS1	Citrullinemia	AR	Worldwide	1 in 1343	99	1 in 134300	•	•		•	•
ATM	Ataxia-Telangiectasia	AR	Worldwide	1 in 423	99	1 in 42300	•		•	•	
ATP13A2	Kufor-Rakeb syndrome and Spastic paraplegia	AR	Worldwide	1 in 900	99	1 in 90000	•			•	
ATP6V1B1	Renal tubular acidosis with deafness	AR	Worldwide	1 in 866	99	1 in 86600	•			•	
ATP7A	Menkes disease	XLR	Worldwide	1 in 175000	99	1 in 17500000	•				
ATP7B	Wilson disease	AR	Worldwide	1 in 97	99	1 in 9700	•	•	•	•	•
ATRX	Alpha-thalassemia intellectual disability syndrome	XLR	Worldwide	1 in 100000	99	1 in 10000000	•				
AVPR2	Nephrogenic diabetes insipidus	XLR	Worldwide	1 in 144000	97	1 in 4800000	•				
B9D1	Meckel syndrome	AR	Worldwide	1 in 5400	99	1 in 540000	•			•	
B9D2	Meckel syndrome	AR	Worldwide	1 in 5400	99	1 in 540000	•			•	
BBS1	Bardet-Biedl syndrome	AR	Worldwide	1 in 520	99	1 in 52000	•	•		•	•

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BBS10	Bardet-Biedl syndrome	AR	Worldwide	1 in 562	99	1 in 56200	•	•		•	•
BBS12	Bardet-Biedl syndrome	AR	Worldwide	1 in 1230	99	1 in 123000	•	•		•	•
BBS2	Bardet-Biedl syndrome and Retinitis pigmentosa	AR	Worldwide	1 in 870	99	1 in 87000	•	•	•	•	•
BBS4	Bardet-Biedl syndrome	AR	Worldwide	1 in 768	99	1 in 76800	•			•	
BBS5	Bardet-Biedl syndrome	AR	Worldwide	1 in 919	99	1 in 91900	•			•	
BBS7	Bardet-Biedl syndrome	AR	Worldwide	1 in 873	99	1 in 87300	•			•	
BBS9	Bardet-Biedl syndrome	AR	Worldwide	1 in 959	99	1 in 95900	•			•	
BCKDHA	Maple syrup urine disease	AR	Worldwide	1 in 1002	99	1 in 100200	•		•	•	
BCKDHB	Maple syrup urine disease	AR	Worldwide	1 in 475	99	1 in 47500	•	•	•	•	•
BCS1L	GRACILE syndrome and other BCS1L related disorders	AR	Worldwide	1 in 720	99	1 in 72000	•			•	
BLM	Bloom syndrome	AR	Worldwide	1 in 632	99	1 in 63200	•	•	•	•	•
BRAT1	Lethal neonatal rigidity and multifocal seizure syndrome	AR	Worldwide	1 in 540	99	1 in 54000	•			•	
BSND	Bartter syndrome	AR	Worldwide	1 in 866	99	1 in 86600	•			•	
BTD	Biotinidase deficiency	AR	Worldwide	1 in 642	99	1 in 64200	•	•		•	•
CANT1	Desbuquois dysplasia and Epiphyseal dysplasia, multiple	AR	Worldwide	1 in 1170	99	1 in 117000	•			•	
CAPN3	Limb-girdle muscular dystrophy	AR	Worldwide	1 in 112	99	1 in 11200	•			•	
CASQ2	Catecholaminergic polymorphic ventricular tachycardia (CPVT)	AR	Worldwide	1 in 1661	99	1 in 166140	•	•		•	•
CBS	Homocystinuria due to cystathionine beta-synthase deficiency	AR	Worldwide	1 in	99	1 in 41100	•	•		•	•
CC2D2A Excluded from the analysis: NM_020785, exon 7	Joubert syndrome, Meckel syndrome and other CC2D2A related disorders	AR	Worldwide	1 in 389	99	1 in 38900	•	•		•	•
CCDC88C	Congenital hydrocephalus type 1	AR	Worldwide	1 in 2236	99	1 in 223600	•	•		•	•
CD40	Hyper-IgM syndrome	AR	Worldwide	1 in 21600	99	1 in 2160000	•			•	
CD40LG	Hyper-IgM syndrome	XLR	Worldwide	1 in 36000	97	1 in 1200000	•				
CDH23	Usher syndrome type 1D	AR	Worldwide	1 in 970	99	1 in 97000	•			•	

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CENPJ	Primary microcephaly and Seckel syndrome	AR	Worldwide	1 in 540	99	1 in 54000	•			•	
CEP290[#]	Leber congenital amaurosis 10 and other CEP290 related ciliopathies	AR	Worldwide	1 in 84	99	1 in 8400	•	•		•	•
CERKL	Retinitis pigmentosa	AR	Worldwide	1 in 194	99	1 in 19400	•			•	
CFTR	Cystic fibrosis	AR	Worldwide	1 in 42	99	1 in 4200	•	•	•	•	•
CHAT	Congenital myasthenic syndrome	AR	Worldwide	1 in 939	99	1 in 93870	•			•	
CHM Excluded from the analysis: NM_001145414, exon 5	Choroideremia	XLR	Worldwide	1 in 37500	96	1 in 937500	•				
CHRNE	Myasthenic syndrome	AR	Worldwide	1 in 302	99	1 in 30200	•	•		•	•
CIITA	Bare lymphocyte syndrome	AR	Worldwide	1 in 1224	99	1 in 122400	•			•	
CLCN1	Myotonia congenita	AR	Worldwide	1 in 632	99	1 in 63200	•	•		•	•
CLN3	Neuronal ceroid lipofuscinosis type 3	AR	Worldwide	1 in 760	99	1 in 76000	•	•		•	•
CLN5	Neuronal ceroid lipofuscinosis type 5	AR	Worldwide	1 in 1021	99	1 in 102100	•			•	
CLN6	Neuronal ceroid lipofuscinosis type 6	AR	Worldwide	1 in 4355	99	1 in 435500	•			•	
CLN8	Neuronal ceroid lipofuscinosis type 8	AR	Worldwide	1 in 1896	99	1 in 189600	•			•	
CLRN1	Usher sydnrome type 3A and Retinitis pigmentosa	AR	Worldwide	1 in 970	99	1 in 97000	•	•	•	•	•
CNGA3	Achromatopsia	AR	Worldwide	1 in 157	99	1 in 15660	•			•	
CNGB3	Achromatopsia and Juvenile macular degeneration	AR	Worldwide	1 in 978	99	1 in 97800	•	•		•	•
COL27A1	Steel syndrome	AR	Worldwide	1 in 500	99	1 in 50000	•			•	
COL4A3	Alport syndrome	AR	Worldwide	1 in 500	99	1 in 50000	•		•	•	
COL4A4	Alport syndrome	AR	Worldwide	1 in 707	99	1 in 70700	•			•	
COL4A5	Alport syndrome	XLD/XLR	Worldwide	1 in 30000	98	1 in 1500000	•			•	
COL7A1	Epidermolysis bullosa dystrophica	AR	Worldwide	1 in 152	99	1 in 15200	•	•		•	•
COLQ	Congenital myasthenic syndrome	AR	Worldwide	1 in 939	99	1 in 93870	•			•	
CPS1	Carbamoylphosphate synthetase I deficiency	AR	Worldwide	1 in 4877	99	1 in 487700	•			•	

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CPT1A	Carnitine palmitoyltransferase deficiency	AR	Worldwide	1 in 3115	99	1 in 311500	•			•	
CPT2	Carnitine palmitoyltransferase II deficiency	AR	Worldwide	1 in 1157	99	1 in 115700	•	•	•	•	•
CRB1	Leber congenital amaurosis, Pigmented paravenous chorioretinal atrophy and Retinitis pigmentosa	AD/AR	Worldwide	1 in 112	99	1 in 11200	•			•	
CRPPA	Muscular dystrophy-dystroglycanopathy		Worldwide	1 in 2842	99	1 in 284220	•			•	
CTNS	Cystinosis	AR	Worldwide	1 in 1179	99	1 in 117900	•		•	•	
CTSD	Neuronal ceroid lipofuscinosis	AR	Worldwide	1 in 7200	99	1 in 720000	•			•	
CTSF	Neuronal ceroid lipofuscinosis	AR	Worldwide	1 in 1080	93	1 in 15429	•			•	
CTSK	Pycnodysostosis	AR	Worldwide	1 in 3382	99	1 in 338200	•			•	
CYBA	Chronic granulomatous disease	AR	Worldwide	1 in 866	99	1 in 86600	•			•	
CYBB	Chronic granulomatous disease	XLR	Worldwide	1 in 150000	99	1 in 15000000	•				
CYP11B1[#]	Congenital adrenal hyperplasia due to 11-Beta-Hydroxylase Deficiency	AR	Worldwide	1 in 224	92	1 in 2800	•			•	
CYP11B2	Corticosterone methyloxidase type I deficiency and Corticosterone methyloxidase type II deficiency	AR	Worldwide	1 in 1420	99	1 in 142000	•			•	
CYP17A1	Congenital adrenal hyperplasia due to 17-Alpha-hydroxylase deficiency	AR	Worldwide	1 in 112	99	1 in 11200	•			•	
CYP19A1	Aromatase deficiency	AR	Worldwide	1 in 707	99	1 in 70700	•			•	
CYP1B1	Primary congenital glaucoma	AR	Worldwide	1 in 235	97	1 in 7830	•			•	
CYP21A2[#]	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency|Hyperandrogenism, nonclassic , due to 21-hydroxylase deficiency	AR	Worldwide	1 in 61	75	1 in 244	•	•		•	•
CYP27A1	Cerebrotendinous xanthomatosis	AR	Worldwide	1 in 402	99	1 in 40200	•	•		•	•
CYP27B1	Vitamin D-dependent rickets	AR	Worldwide	1 in 578	99	1 in 57800	•	•		•	•
CYP7B1	Spastic paraplegia type 5A	AR	Worldwide	1 in 470	99	1 in 46980	•			•	
DBT	Maple syrup urine disease	AR	Worldwide	1 in 513	99	1 in 51300	•			•	
DCLRE1C[#]	Severe combined immunodeficiency with sensitivity to ionizing radiation	AR	Worldwide	1 in 1682	98	1 in 84100	•			•	

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DDX11	Warsaw breakage syndrome	AR	Worldwide	1 in 1581	99	1 in 158100	•			•	
DGAT1	Diarrhea	AR	Worldwide	1 in 1350	95	1 in 27000	•			•	
DHCR7	Smith-Lemli-Opitz syndrome	AR	Worldwide	1 in 87	99	1 in 8700	•	•	•	•	•
DHDDS	Retinitis pigmentosa	AR	Worldwide	1 in 2344	99	1 in 234400	•	•	•	•	•
DKC1	Dyskeratosis congenita and Hoyeraal-Hreidarsson syndrome	XLR	Worldwide	1 in 2000000	99	1 in 200000000	•				
DLD	Dihydrolipoyl dehydrogenase deficiency	AR	Worldwide	1 in 1130	99	1 in 113000	•	•	•	•	•
DMD	Duchenne & Becker muscular dystrophy and Dilated cardiomyopathy	XLR/XLD	Worldwide	1 in 2000	99	1 in 200000	•	•			
DNAH5	Ciliary dyskinesia	AR	Worldwide	1 in 178	99	1 in 17800	•		•	•	
DNAI1	Ciliary dyskinesia	AR	Worldwide	1 in 720	99	1 in 72000	•		•	•	
DNAI2	Ciliary dyskinesia	AR	Worldwide	1 in 689	99	1 in 68900	•		•	•	
DOK7	Congenital myasthenic syndrome	AR	Worldwide	1 in 373	95	1 in 7452	•			•	
DYNC2H1	Short -rib thoracic dysplasia with or without polydactyly type 1	AR	Worldwide	1 in 273	99	1 in 27300	•	•		•	•
DYSF	Limb-girdle muscular dystrophy	AR	Worldwide	1 in 470	99	1 in 47000	•			•	
EDA	Hypohidrotic ectodermal dysplasia	XLR	Worldwide	1 in 5700	99	1 in 570000	•				
EIF2AK4	Pulmonary venoocclusive disease	AR	Worldwide	1 in 635	99	1 in 63540	•			•	
EIF2B2	Leukoencephalopathy with vanishing white matter	AR	Worldwide	1 in 783	99	1 in 78300	•			•	
EIF2B5	Leukoencephalopathy with vanishing white matter and Ovarioleukodystrophy	AR	Worldwide	1 in 1472	99	1 in 147200	•			•	
ELP1	Familial dysautonomia	AR	Worldwide	1 in 373	99	1 in 37260	•	•	•	•	•
EMD	Emery-Dreifuss muscular dystrophy	XLR	Worldwide	1 in 300000	99	1 in 30000000	•				
EPG5	Vici syndrome	AR	Worldwide	1 in 864	99	1 in 86400	•			•	
ERCC2	Xeroderma pigmentosum and Photo-sensitive trichothiodystrophy	AR	Worldwide	1 in 167	99	1 in 16700	•	•		•	•
ERCC6#	Cockayne syndrome type B and other ERCC6 related disorders	AR	Worldwide	1 in 723	98	1 in 36150	•			•	
ERCC6L2	Bone marrow failure syndrome 2	AR	Worldwide	1 in 1080	99	1 in 108000	•			•	
ERCC8	Cockayne syndrome	AR	Worldwide	1 in 4730	99	1 in 473000	•			•	

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ESCO2	Roberts syndrome	AR	Worldwide	1 in 978	99	1 in 97800	•			•	
ETFA	Glutaric aciduria type II	AR	Worldwide	1 in 1997	99	1 in 199700	•			•	
ETFB	Glutaric aciduria	AR	Worldwide	1 in 3600	99	1 in 360000	•			•	
ETFDH	Glutaric aciduria	AR	Worldwide	1 in 1422	99	1 in 142200	•			•	
ETHE1	Ethylmalonic encephalopathy	AR	Worldwide	1 in 3214	99	1 in 321400	•			•	
EVC	Ellis-van Creveld syndrome	AR	Worldwide	1 in 1630	96	1 in 40750	•			•	
EVC2	Ellis-van Creveld syndrome	AR	Worldwide	1 in 1230	99	1 in 123000	•	•		•	•
EXOSC3	Pontocerebellar hypoplasia	AR	Worldwide	1 in 514	99	1 in 51390	•			•	
EYS#	Retinitis pigmentosa	AR	Worldwide	1 in 189	99	1 in 18900	•			•	
F11	Factor XI deficiency	AD/AR	Worldwide	1 in 219	99	1 in 21900	•		•	•	
F9	Factor IX deficiency	XLD/XLR	Worldwide	1 in 2000	99	1 in 200000	•	•			
FAH	Tyrosinemia	AR	Worldwide	1 in 371	99	1 in 37100	•	•	•	•	•
FAM161A	Retinitis pigmentosa	AR	Worldwide	1 in 534	99	1 in 53400	•		•	•	
FANCA	Fanconi anemia	AR	Worldwide	1 in 1756	99	1 in 175600	•	•		•	•
FANCC	Fanconi anemia	AR	Worldwide	1 in 623	99	1 in 62300	•	•	•	•	•
FANCE	Fanconi anemia	AR	Worldwide	1 in 1350	99	1 in 135000	•			•	
FANCG	Fanconi anemia	AR	Worldwide	1 in 1736	99	1 in 173600	•			•	
FANCI	Fanconi anemia	AR	Worldwide	1 in 569	99	1 in 56880	•			•	
FANCL	Fanconi anemia	AR	Worldwide	1 in 1800	99	1 in 180000	•			•	
FH	Fumarase deficiency	AR	Worldwide	1 in 970	99	1 in 97000	•			•	
FHL1#	Reducing bod myopathy and Hypertrophic and dilated cardiomyopathy	XL	Worldwide	1 in 1000000	99	1 in 100000000	•			•	
FKRP	Muscular dystrophy-dystroglycanopathy	AR	Worldwide	1 in 350	99	1 in 35000	•	•		•	•
FKTN	Muscular dystrophy-dystroglycanopathy	AR	Worldwide	1 in 724	99	1 in 72400	•	•	•	•	•
FMO3	Trimethylaminuria	AR	Worldwide	1 in 321	99	1 in 32100	•	•		•	•
FMR1	Fragile X syndrome	XLD	Worldwide	1 in 4000 full mutation	99	1 in 400000	•	•	•		
FMR1	Fragile X syndrome	XLD	Worldwide	1 in 201 premutation	99	1 in 20100	•	•	•		
FRAS1	Fraser syndrome	AR	Worldwide	1 in 540	99	1 in 54000	•			•	
G6PC	Glycogen storage disease	AR	Worldwide	1 in 354	99	1 in 35400	•	•	•	•	•
GAA	Glycogen storage disease	AR	Worldwide	1 in 134	99	1 in 13400	•	•	•	•	•
GALC	Krabbe disease	AR	Worldwide	1 in 784	99	1 in 78400	•			•	

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GALE	Galactose epimerase deficiency	AR	Worldwide	1 in 3375	99	1 in 337500	•			•	
GALK1	Galactokinase deficiency	AR	Worldwide	1 in 780	99	1 in 78000	•			•	
GALNS	Mucopolysaccharidosis (Morquio syndrome)	AR	Worldwide	1 in 432	97	1 in 14400	•			•	
GALNT3	Familial hyperphosphatemic tumoral calcinosis	AR	Worldwide	1 in 794	99	1 in 79380	•			•	
GALT	Galactosemia	AR	Worldwide	1 in 183	99	1 in 18300	•	•	•	•	•
GAMT	Guanidinoacetate methyltransferase deficiency	AR	Worldwide	1 in 917	99	1 in 91700	•			•	
GBA#	Gaucher disease	AR	Worldwide	1 in 360	90	1 in 3600	•	•	•	•	•
GBE1	Glycogen storage disease	AR	Worldwide	1 in 316	99	1 in 31600	•	•		•	•
GCDH	Glutaric aciduria	AR	Worldwide	1 in 217	99	1 in 21700	•			•	
GCH1	BH4-deficient hyperphenylalaninemia B	AR	Worldwide	1 in 15428	99	1 in 1542780	•			•	
GCSH	Glycine encephalopathy	AR	Worldwide	1 in 3600	90	1 in 36000	•			•	
GFM1	Combined oxidative phosphorylation deficiency	AR	Worldwide	1 in 1223	99	1 in 122300	•			•	
GFPT1	Congenital myasthenic syndrome	AR	Worldwide	1 in 3086	99	1 in 308610	•			•	
GJB1	Charcot-Marie-Tooth neuropathy	XLD	Worldwide	1 in 5000	99	1 in 500000	•		•	•	
GJB2	Deafness	AD/AR	Worldwide	1 in 37	99	1 in 3700	•	•	•	•	•
GJB6	Deafness	AD/AR	Worldwide	1 in 238	99	1 in 23800	•	•		•	•
GLA	Fabry disease	XLR/XLD	Worldwide	1 in 40000	99	1 in 4000000	•	•		•	•
GLB1	GM1-gangliosidosis|Mucopoly-saccharidosis (Morquio syndrome)	AR	Worldwide	1 in 512	99	1 in 51200	•	•		•	•
GLDC	Glycine encephalopathy	AR	Worldwide	1 in 722	99	1 in 72200	•			•	
GLE1	Lethal congenital contracture syndrome	AR	Worldwide	1 in 2341	99	1 in 234100	•			•	
GNE	Nonaka myopathy	AR	Worldwide	1 in 236	99	1 in 23600	•			•	
GNPAT	Rhizomelic chondrodysplasia punctata	AR	Worldwide	1 in 3600	99	1 in 360000	•			•	
GNPTAB	Mucolipidosis	AR	Worldwide	1 in 213	99	1 in 21300	•	•		•	•
GNPTG	Mucolipidosis	AR	Worldwide	1 in 1258	99	1 in 125800	•			•	

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GNS	Mucopolysaccharidosis (Sanfilippo syndrome)	AR	Worldwide	1 in 5368	99	1 in 536800	•			•	
GORAB	Geroderma osteodysplasticum	AR	Worldwide	1 in 2348	99	1 in 234810	•			•	
GP9	Bernard-Soulier syndrome	AR	Worldwide	1 in 2144	99	1 in 214400	•			•	
GRHPR	Primary hyperoxaluria type 2	AR	Worldwide	1 in 866	99	1 in 86600	•			•	
GRIP1	Fraser syndrome	AR	Worldwide	1 in 489	99	1 in 48900	•			•	
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency	AR	Worldwide	1 in 6353	99	1 in 635310	•			•	
HADHA	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	AR	Worldwide	1 in 416	99	1 in 41600	•			•	
HADHB	Trifunctional protein deficiency	AR	Worldwide	1 in 1113	99	1 in 111330	•			•	
HAMP	Juvenile hemochromatosis	AR	Worldwide	1 in 6750	99	1 in 675000	•			•	
HAX1	Severe congenital neutropenia	AR	Worldwide	1 in 3785	99	1 in 378500	•			•	
HBA1[#]	Alpha-thalassemia (Hemoglobin Bart syndrome / Hemoglobin H disease)	AR/Di-genic	Worldwide	1 in 50	90	1 in 500	•	•	•	•	•
HBA2[#]	Alpha-thalassemia (Hemoglobin Bart syndrome / Hemoglobin H disease)	AR/Di-genic	Worldwide	1 in 50	90	1 in 500	•	•	•	•	•
HBB	Beta-thalassemia and Other Thalas-semias/Hemoglobinopathies	AD/AR/ Digenic	Worldwide	1 in 116	99	1 in 11600	•	•	•	•	•
HEXA	Tay-Sachs disease and Hexosaminidase A deficiency	AR	Worldwide	1 in 423	99	1 in 42300	•	•	•	•	•
HEXB	Sandhoff disease	AR	Worldwide	1 in 587	99	1 in 58700	•			•	
HGD	Alkaptonuria	AR	Worldwide	1 in 500	99	1 in 50000	•			•	
HGSNAT	Mucopolysaccharidosis (Sanfilippo syndrome) and Retinitis pigmentosa	AR	Worldwide	1 in 1023	97	1 in 34100	•			•	
HJV	Juvenile hemochromatosis	AR	Worldwide	1 in 1080	99	1 in 108000	•			•	
HLCS	Holocarboxylase synthetase deficiency	AR	Worldwide	1 in 912	99	1 in 91200	•			•	
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR	Worldwide	1 in 2240	99	1 in 224000	•			•	
HOGA1	Primary hyperoxaluria type 3	AR	Worldwide	1 in 866	99	1 in 86600	•		•	•	
HPRT1	Kelley-Seegmiller syndrome and Lesch-Nyhan syndrome	XLR	Worldwide	1 in 760000	97	1 in 25333333	•				

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HPS1[#]	Hermansky-Pudlak syndrome	AR	Worldwide	1 in 500	98	1 in 25000	•	•		•	•
HPS3	Hermansky-Pudlak syndrome	AR	Worldwide	1 in 1581	99	1 in 158100	•	•	•	•	•
HPS4	Hermansky-Pudlak syndrome	AR	Worldwide	1 in 1367	99	1 in 136710	•			•	
HPS5	Hermansky-Pudlak syndrome	AR	Worldwide	1 in 1350	99	1 in 135000	•			•	
HPS6	Hermansky-Pudlak syndrome	AR	Worldwide	1 in 1636	99	1 in 163620	•			•	
HSD17B3	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation	AR	Worldwide	1 in 450	99	1 in 45000	•			•	
HSD17B4	D-bifunctional protein deficiency and Perrault syndrome	AR	Worldwide	1 in 899	99	1 in 89900	•			•	
HSD3B2	3-beta-hydroxysteroid dehydrogenase II deficiency	AR	Worldwide	1 in 1922	99	1 in 192200	•			•	
HYLS1	Hydrolethalus syndrome	AR	Worldwide	1 in 1197	99	1 in 119700	•			•	
IDS[#]	Mucopolysaccharidosis type 2	XLR	Worldwide	1 in 206	90	1 in 2060	•				
IDUA	Mucopolysaccharidosis	AR	Worldwide	1 in 158	99	1 in 15800	•	•		•	•
IL2RG	Combined immunodeficiency	XLR	Worldwide	1 in 100000	99	1 in 10000000	•				
IL7R	Severe combined immunodeficiency	AR	Worldwide	1 in 1440	99	1 in 144000	•			•	
IQCB1	Senior-Loken syndrome	AR	Worldwide	1 in 628	99	1 in 62820	•			•	
IVD	Isovaleric acidemia	AR	Worldwide	1 in 565	99	1 in 56500	•	•		•	•
JAK3	Severe combined immunodeficiency	AR	Worldwide	1 in 2160	99	1 in 216000	•			•	
KCNJ1	Bartter syndrome, antenatal	AR	Worldwide	1 in 720	99	1 in 72000	•			•	
KCTD7	Epilepsy, progressive myoclonic	AR	Worldwide	1 in 3600	99	1 in 360000	•			•	
KIAA0586 Excluded from the analysis: NM_001244189, exons 6 and 33	Joubert syndrome and Short rib thoracic dysplasia with polydactyly	AR	Worldwide	1 in 124	95	1 in 2484	•	•		•	•
KIF14	Meckel syndrome 12	AR	Worldwide	1 in 1800	99	1 in 180000	•			•	
L1CAM	Hydrocephalus due to congenital stenosis of aqueduct of Sylvius and Other L1CAM related disorders	XLR	Worldwide	1 in 16000	99	1 in 1600000	•	•			
LAMA2	LAMA2 related muscular dystrophy	AR	Worldwide	1 in 857	99	1 in 85700	•			•	

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LAMA3	Junctional epidermolysis bullosa	AR	Worldwide	1 in 1856	99	1 in 185600	•			•	
LAMB3	Junctional epidermolysis bullosa	AR	Worldwide	1 in 673	99	1 in 67300	•			•	
LAMC2	Junctional epidermolysis bullosa	AR	Worldwide	1 in 2112	99	1 in 211200	•			•	
LARGE1	Muscular dystrophy-dystroglycanopathy	AR	Worldwide	1 in 3724	85	1 in 24828	•			•	
LCA5	Leber congenital amaurosis	AR	Worldwide	1 in 1417	99	1 in 141700	•			•	
LDLR	Familial hypercholesterolemia	AD/AR	Worldwide	1 in 120	99	1 in 12000	•			•	
LHX3	Combined pituitary hormone deficiency	AR	Worldwide	1 in 3725	99	1 in 372500	•			•	
LIFR	Stuve-Wiedemann dysplasia	AR	Worldwide	1 in 2153	99	1 in 215300	•			•	
LIG4	LIG4 syndrome (Severe combined immunodeficiency with sensitivity to ionizing radiation)	AR	Worldwide	1 in 554	99	1 in 55350	•			•	
LIPA	Cholesterol ester storage disease and Wolman disease	AR	Worldwide	1 in 614	99	1 in 61400	•			•	
LIPH	Hypotrichosis 7	AR	Worldwide	1 in 720	99	1 in 72000	•			•	
LOXHD1	Deafness	AR	Worldwide	1 in 513	99	1 in 51300	•		•	•	
LRP2	Donnai-Barrow syndrome	AR	Worldwide	1 in 462	99	1 in 46200	•	•		•	•
LRPPRC	Leigh syndrome French-Canadian type	AR	Worldwide	1 in 1347	99	1 in 134700	•			•	
LYST	Chediak-Higashi syndrome	AR	Worldwide	1 in 1440	99	1 in 144000	•			•	
MAN2B1	Alpha mannosidosis	AR	Worldwide	1 in 734	99	1 in 73400	•			•	
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	AR	Worldwide	1 in 628	99	1 in 62800	•			•	
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	AR	Worldwide	1 in 754	99	1 in 75400	•	•		•	•
MCOLN1	Mucolipidosis	AR	Worldwide	1 in 1785	99	1 in 178500	•	•	•	•	•
MCPH1 Excluded from the analysis: NM_001322042, exon 14	Microcephaly	AR	Worldwide	1 in 623	97	1 in 20767	•	•		•	•
MED17	Postnatal progressive microcephaly with seizures and brain atrophy	AR	Worldwide	1 in 1988	99	1 in 198800	•			•	
MEFV	Familial Mediterranean fever	AR	Worldwide	1 in 145	97	1 in 4830	•		•	•	
MESP2	Spondylocostal dysostosis 2	AR	Worldwide	1 in 2247	99	1 in 224700	•			•	

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MFSD8	Neuronal ceroid lipofuscinosis	AR	Worldwide	1 in 1784	99	1 in 178400	•	•		•	•
MID1[#]	Opitz GBBB syndrome	XLR	Worldwide	1 in 56250	96	1 in 1406250	•	•			
MKKS	Bardet-Biedl syndrome and McKusick-Kaufman syndrome	AR	Worldwide	1 in 1125	99	1 in 112500	•			•	
MKS1	Bardet-Biedl syndrome and Meckel syndrome	AR	Worldwide	1 in 632	97	1 in 21067	•			•	
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR	Worldwide	1 in 1021	99	1 in 102100	•			•	
MMAA	Methylmalonic acidemia	AR	Worldwide	1 in 1082	99	1 in 108200	•			•	
MMAB	Methylmalonic acidemia	AR	Worldwide	1 in 1866	99	1 in 186600	•			•	
MMACHC	Methylmalonic aciduria and homocystinuria	AR	Worldwide	1 in 207	99	1 in 20700	•	•		•	
MMADHC	Methylmalonic aciduria and homocystinuria	AR	Worldwide	1 in 3784	99	1 in 378400	•			•	•
MMUT	Methylmalonic acidemia due to methylmalonyl-CoA mutase deficiency	AR	Worldwide	1 in 257	99	1 in 25740	•			•	
MPI	Congenital disorder of glycosylation	AR	Worldwide	1 in 1368	99	1 in 136800	•			•	
MPL	Congenital megakaryocytic thrombocytopenia	AR	Worldwide	1 in 812	99	1 in 81200	•		•	•	
MPV17	Mitochondrial DNA depletion syndrome	AR	Worldwide	1 in 1216	99	1 in 121600	•			•	
MRE11	Ataxia-telangiectasia-like disorder-1	AR	Worldwide	1 in 1029	99	1 in 102870	•			•	
MTHFR	Homocystinuria due to MTHFR deficiency	AR	Worldwide	1 in 1261	99	1 in 126100	•			•	
MTM1	Centronuclear myopathy	XLR	Worldwide	1 in 25000	98	1 in 1250000	•				
MTTP	Abetalipoproteinemia	AR	Worldwide	1 in 2540	99	1 in 254000	•		•	•	
MVK	Mevalonic aciduria	AR	Worldwide	1 in 374	99	1 in 37400	•	•		•	•
MYO15A	Deafness	AR	Worldwide	1 in 163	99	1 in 16290	•			•	
MYO7A	Usher syndrome type I	AR	Worldwide	1 in 384	99	1 in 38400	•			•	
NAGLU	Mucopolysaccharidosis (Sanfilippo syndrome)	AR	Worldwide	1 in 286	99	1 in 28600	•			•	
NAGS	N-acetylglutamate synthase deficiency	AR	Worldwide	1 in 1837	99	1 in 183700	•			•	
NBN	Nijmegen breakage syndrome	AR	Worldwide	1 in 1623	99	1 in 162300	•			•	

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NCF2	Chronic granulomatous disease	AR	Worldwide	1 in 2038	99	1 in 203760	•			•	
NDRG1	Charcot-Marie-Tooth disease	AR	Worldwide	1 in 7234	99	1 in 723400	•			•	
NDUFAF5	Mitochondrial complex I deficiency	AR	Worldwide	1 in 1423	99	1 in 142300	•			•	
NDUFAF6	Leigh syndrome	AR	Worldwide	1 in 1661	99	1 in 166140	•			•	
NDUFS4	Leigh syndrome due to mitochondrial complex I deficiency	AR	Worldwide	1 in 1589	99	1 in 158900	•			•	
NDUFS6	Mitochondrial complex I deficiency	AR	Worldwide	1 in 5468	99	1 in 546800	•		•	•	
NEB#	Nemaline myopathy	AR	Worldwide	1 in 1134	86	1 in 8100	•	•	•	•	•
NGLY1	Congenital disorder of deglycosylation	AR	Worldwide	1 in 939	99	1 in 93870	•			•	
NPC1	Niemann-Pick disease	AR	Worldwide	1 in 487	98	1 in 24350	•			•	
NPC2	Niemann-pick disease	AR	Worldwide	1 in 3855	99	1 in 385500	•			•	
NPHP1	Joubert syndrome, Nephronophthisis and Senior-Loken syndrome	AR	Worldwide	1 in 235	99	1 in 23490	•			•	
NPHP3	Meckel syndrome, Nephronophthisis and Renal-hepatic-pancreatic dysplasia	AR	Worldwide	1 in 376	99	1 in 37620	•			•	
NPHS1	Nephrotic syndrome	AR	Worldwide	1 in 152	99	1 in 15200	•	•		•	•
NPHS2	Nephrotic syndrome	AR	Worldwide	1 in 987	99	1 in 98700	•			•	
NR0B1	Congenital adrenal hypoplasia	XLR	Worldwide	1 in 60000	99	1 in 6000000	•	•			
NR2E3	Enhanced S-cone syndrome and Retinitis pigmentosa	AD/AR	Worldwide	1 in 894	99	1 in 89400	•		•	•	
NTRK1	Congenital insensitivity to pain with anhidrosis	AR	Worldwide	1 in 1196	99	1 in 119600	•			•	
OAT	Gyrate atrophy of choroid and retina	AR	Worldwide	1 in 892	99	1 in 89200	•			•	
OCA2	Albinism	AR	Worldwide	1 in 164	99	1 in 16400	•	•		•	•
OPA3	3-methylglutaconic aciduria and Optic atrophy	AD/AR	Worldwide	1 in 6258	99	1 in 625800	•			•	
ORC4	Meier-Gorlin syndrome (Ear-patel-la-short stature syndrome)	AR	Worldwide	1 in 1543	99	1 in 154260	•			•	
OTC	Ornithine transcarbamylase deficiency	XLR/XLD	Worldwide	1 in 30000	98	1 in 1500000	•	•			
OTOF	Deafness	AR	Worldwide	1 in 410	99	1 in 41040	•			•	
PAH	Phenylketonuria	AR	Worldwide	1 in 62	99	1 in 6200	•	•	•	•	•

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PC	Pyruvate carboxylase deficiency	AR	Worldwide	1 in 5966	99	1 in 596600	•			•	
PCCA	Propionic acidemia	AR	Worldwide	1 in 1322	99	1 in 132200	•	•		•	•
PCCB Excluded from the analysis: NM_001178014, exon 4	Propionic acidemia	AR	Worldwide	1 in 972	99	1 in 97200	•	•		•	•
PCDH15	Usher syndrome type 1D	AR	Worldwide	1 in 874	99	1 in 87400	•	•	•	•	•
PCNT	Microcephalic osteodysplastic primordial dwarfism	AR	Worldwide	1 in 360	99	1 in 36000	•			•	
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	XLD	Worldwide	1 in 50000	99	1 in 5000000	•			•	
PDHB	Pyruvate dehydrogensae E1-beta deficiency	AR	Worldwide	1 in 3874	99	1 in 387400	•			•	
PEPD	Prolidase deficiency	AR	Worldwide	1 in 857	99	1 in 85680	•			•	
PET100	Mitochondrial complex IV deficiency	AR	Worldwide	1 in 9000	99	1 in 900000	•			•	
PEX1	Zellweger spectrum disorder	AR	Worldwide	1 in 673	99	1 in 67300	•	•		•	•
PEX10	Zellweger spectrum disorder	AR	Worldwide	1 in 4213	99	1 in 421300	•			•	
PEX12	Zellweger spectrum disorder	AR	Worldwide	1 in 964	99	1 in 96400	•			•	
PEX2	Zellweger spectrum disorder	AR	Worldwide	1 in 2145	99	1 in 214500	•		•	•	
PEX26	Peroxisome biogenesis disorder	AR	Worldwide	1 in 1688	93	1 in 24107	•			•	
PEX6	Zellweger spectrum disorder	AR	Worldwide	1 in 412	99	1 in 41200	•	•	•	•	•
PEX7	Refsum disease and Rhizomelic CDP type 1	AR	Worldwide	1 in 563	99	1 in 56300	•	•		•	•
PFKM	Glycogen storage disease	AR	Worldwide	1 in 1358	99	1 in 135800	•		•	•	
PHGDH	Neu-Laxova syndrome 1	AR	Worldwide	1 in 1463	99	1 in 146300	•		•	•	
PHKB	Glycogen storage disease	AR	Worldwide	1 in 590	99	1 in 59040	•			•	
PHYH	Refsum disease	AR	Worldwide	1 in 500	99	1 in 50000	•			•	
PKHD1	Polycystic kidney disease	AR	Worldwide	1 in 697	99	1 in 69700	•	•	•	•	•
PLP1	Pelizaeus-Merzbacher disease and Spastic paraplegia	XLR	Worldwide	1 in 200000	99	1 in 20000000	•	•			
PMM2	Congenital disorder of glycosylation	AR	Worldwide	1 in 114	99	1 in 11400	•	•	•	•	•

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PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	AR	Worldwide	1 in 1200	99	1 in 119970	•			•	
POLG	POLG-related ataxia neuropathy spectrum disorders	AR	Worldwide	1 in 365	99	1 in 36500	•	•		•	•
POMGNT1	Muscular dystrophy-dystroglycanopathy	AR	Worldwide	1 in 785	99	1 in 78500	•			•	
POMT1	Muscular dystrophy-dystroglycanopathy	AR	Worldwide	1 in 581	99	1 in 58050	•			•	
POMT2	Muscular dystrophy-dystroglycanopathy	AR	Worldwide	1 in 1080	99	1 in 108000	•			•	
POR	POR deficiency	AR	Worldwide	1 in 450	99	1 in 45000	•			•	
PPT1	Neuronal ceroid lipofuscinosis	AR	Worldwide	1 in 346	99	1 in 34600	•	•		•	•
PRF1	Hemophagocytic lymphohistiocytosis	AR	Worldwide	1 in 1657	99	1 in 165700	•	•		•	•
PROP1	Combined pituitary hormone deficiency	AR	Worldwide	1 in 2868	99	1 in 286800	•			•	
PRPS1[#]	PRPS1 related disorders	XLR/XLD	Worldwide	1 in 50000	99	1 in 5000000	•			•	
PSAP	PSAP related disorders	AR	Worldwide	1 in 5687	99	1 in 568700	•			•	
PTS	Hyperphenylalaninemia, BH4-deficient	AR	Worldwide	1 in 917	99	1 in 91700	•			•	
PUS1	Mitochondrial myopathy and sideroblastic anemia	AR	Worldwide	1 in 6712	99	1 in 671200	•			•	
PYGL	Glycogen storage disease	AR	Worldwide	1 in 662	99	1 in 66240	•			•	
PYGM	Glycogen storage disease	AR	Worldwide	1 in 354	99	1 in 35400	•			•	
QDPR	BH4-deficient hyperphenylalaninemia C	AR	Worldwide	1 in 3600	97	1 in 120000	•			•	
RAB23	Carpenter syndrome 1	AR	Worldwide	1 in 1366	99	1 in 136600	•			•	
RAG1	Severe combined immunodeficiency	AR	Worldwide	1 in 392	99	1 in 39240	•	•		•	•
RAG2	Combined cellular and humoral immune defects with granulomas and Omenn syndrome	AR	Worldwide	1 in 1825	99	1 in 182500	•			•	
RAPSN	Congenital myasthenic syndrome	AR	Worldwide	1 in 2389	99	1 in 238900	•			•	
RARS2	Pontocerebellar hypoplasia	AR	Worldwide	1 in 812	99	1 in 81200	•			•	
RDH12	Leber congenital amaurosis and Retinitis pigmentosa	AD/AR	Worldwide	1 in 623	99	1 in 62300	•			•	
RECQL4	Rothmund-Thomson syndrome	AR	Worldwide	1 in 207	98	1 in 10350	•			•	
RMRP	Cartilage-hair hypoplasia	AR	Worldwide	1 in 812	98	1 in 40600	•	•		•	•

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RNASEH2A	Aicardi-Goutières syndrome	AR	Worldwide	1 in 3086	99	1 in 308610	•			•	
RNASEH2B	Aicardi-Goutières syndrome	AR	Worldwide	1 in 416	99	1 in 41600	•	•		•	•
RNASEH2C	Aicardi-Goutières syndrome	AR	Worldwide	1 in 4320	95	1 in 86400	•			•	
RPE65	Leber congenital amaurosis and Retinitis pigmentosa	AD/AR	Worldwide	1 in 589	99	1 in 58900	•			•	
RPGR	Retinitis pigmentosa	XL	Worldwide	1 in 11250	99	1 in 1125000	•	•		•	•
RPGRIP1	Leber congenital amaurosis	AR	Worldwide	1 in 675	99	1 in 67500	•			•	
RPGRIP1L Excluded from the analysis: NM_015272, exon 23	Joubert syndrome, Meckel syndrome and Other ciliopathy syndromes	AR	Worldwide	1 in 947	98	1 in 47350	•			•	
RS1	Retinoschisis	XLR	Worldwide	1 in 10000	99	1 in 1000000	•	•			
RTEL1	Dyskeratosis congenita and Pulmonary fibrosis	AD/AR	Worldwide	1 in 1258	99	1 in 125800	•		•	•	
SACS	Spastic ataxia, Charlevoix-Saguenay	AR	Worldwide	1 in 1473	99	1 in 147300	•			•	
SAMHD1	Aicardi-Goutières syndrome	AR	Worldwide	1 in 1469	99	1 in 146900	•			•	
SCO2	Fatal infantile cardioencephalomyopathy due to cytochrome c oxidase deficiency	AR	Worldwide	1 in 728	99	1 in 72800	•	•		•	•
SEPSECS	Pontocerebellar hypoplasia type 2D	AR	Worldwide	1 in 975	99	1 in 97500	•			•	
SGCA	Limb-girdle muscular dystrophy	AR	Worldwide	1 in 517	99	1 in 51700	•			•	
SGCB	Limb-girdle muscular dystrophy	AR	Worldwide	1 in 2069	99	1 in 206900	•			•	
SGCD	Limb-girdle muscular dystrophy	AR	Worldwide	1 in 4269	99	1 in 426900	•			•	
SGCG	Limb-girdle muscular dystrophy	AR	Worldwide	1 in 2781	99	1 in 278100	•			•	
SGSH	Mucopolysaccharidosis (Sanfilippo syndrome)	AR	Worldwide	1 in 791	99	1 in 79100	•			•	
SLC12A1	Antenatal Bartter syndrome	AR	Worldwide	1 in 900	99	1 in 90000	•			•	
SLC12A3	Gitelman syndrome	AR	Worldwide	1 in 364	99	1 in 36400	•			•	
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy (Andermann syndrome)	AR	Worldwide	1 in 4476	99	1 in 447600	•			•	
SLC17A5	Salla disease	AR	Worldwide	1 in 899	99	1 in 89900	•			•	

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SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	AR	Worldwide	1 in 2700	99	1 in 270000	•			•	
SLC19A3	Thiamine metabolism dysfunction syndrome	AR	Worldwide	1 in 1165	99	1 in 116500	•	•		•	•
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	AR	Worldwide	1 in 2785	99	1 in 278500	•			•	
SLC22A5	Primary carnitine deficiency	AR	Worldwide	1 in 365	98	1 in 18250	•			•	
SLC25A13	Citrin deficiency	AR	Worldwide	1 in 712	99	1 in 71200	•			•	
SLC25A15[#]	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR	Worldwide	1 in 2144	99	1 in 214400	•			•	
SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR	Worldwide	1 in 1137	99	1 in 113670	•			•	
SLC26A2	SLC26A2 related skeletal dysplasias	AR	Worldwide	1 in 356	99	1 in 35600	•	•		•	•
SLC26A3	Congenital Secretory Chloride Diarrhea 1	AR	Worldwide	1 in 491	99	1 in 49050	•			•	
SLC26A4	Pendred syndrome	AR	Worldwide	1 in 128	99	1 in 12800	•	•		•	•
SLC35A2	Congenital disorder of glycosylation	XL	Worldwide	1 in 72000	99	1 in 7200000	•			•	
SLC35A3	Arthrogryposis, impaired intellectual development, and seizures	AR	Worldwide	1 in 4922	99	1 in 492200	•		•	•	
SLC37A4	Glycogen storage disease	AR	Worldwide	1 in 1088	98	1 in 54400	•	•		•	•
SLC39A4	Acrodermatitis enteropathica	AR	Worldwide	1 in 755	99	1 in 75500	•			•	
SLC6A8[#]	Creatine deficiency syndrome	XLR	Worldwide	1 in 50000	97	1 in 1666667	•	•			
SLC7A7	Lysinuric protein intolerance	AR	Worldwide	1 in 827	98	1 in 41350	•			•	
SMARCAL1	Schimke immuno-osseous dysplasia	AR	Worldwide	1 in 1127	99	1 in 112700	•			•	

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SMN1# *Analysis includes only SMN1 copy number analysis, SNV's are not tested in this gene. "Silent" carriers of SMA cannot be detected, i.e., individuals with two copies of SMN1 in one allele, and zero copies in the other allele.	Spinal muscular atrophy	AR	Worldwide	1 in 55	95	1 in 1100	•	•	•	•	•
SMPD1	Niemann-Pick disease	AR	Worldwide	1 in 416	99	1 in 41600	•	•	•	•	•
SPG11	Spastic paraplegia and Amyotrophic lateral sclerosis	AR	Worldwide	1 in 270	97	1 in 9000	•			•	
SPG7	Spastic paraplegia	AR	Worldwide	1 in 232	99	1 in 23220	•			•	
SPINK5	Netherton syndrome	AR	Worldwide	1 in 1581	99	1 in 158100	•			•	
SPR	Sepiapterin reductase deficiency (Dopa-responsive dystonia)	AR	Worldwide	1 in 1256	95	1 in 25110	•			•	
SRD5A2	Steroid 5-alpha-reductase 2 deficiency	AR	Worldwide	1 in 282	99	1 in 28170	•			•	
ST3GAL5	Ganglioside GM3 synthase deficiency	AR	Worldwide	1 in 1137	92	1 in 14209	•			•	
STAR	Lipoid adrenal hyperplasia	AR	Worldwide	1 in 2899	99	1 in 289900	•			•	
STS	Steroid sulfatase deficiency	XLR	Worldwide	1 in 4500	99	1 in 450000	•				
STXBP2	Familial hemophagocytic lymphohistiocytosis	AR	Worldwide	1 in 720	99	1 in 72000	•			•	
SUMF1	Multiple sulfatase deficiency	AR	Worldwide	1 in 1694	99	1 in 169400	•			•	
SURF1	Charcot-Marie-Tooth disease|Leigh syndrome	AR	Worldwide	1 in 635	99	1 in 63540	•		•	•	
SYNE4	Deafness	AR	Worldwide	1 in 2400	99	1 in 240030	•			•	
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration (MECRCN)	AR	Worldwide	1 in 1440	99	1 in 144000	•			•	
TAT	Tyrosinemia type II	AR	Worldwide	1 in 2354	99	1 in 235400	•			•	

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TBCE	Progressive encephalopathy with amyotrophy and optic atrophy (PEAMO)	AR	Worldwide	1 in 2400	99	1 in 240030	•			•	
TCIRG1	Osteopetrosis, severe neonatal or infantile forms (OPTB1)	AR	Worldwide	1 in 1285	99	1 in 128500	•		•	•	
TCTN2	Joubert syndrome and Meckel syndrome	AR	Worldwide	1 in 3652	99	1 in 365200	•			•	
TECPR2	Spastic paraplegia 49	AR	Worldwide	1 in 2844	99	1 in 284400	•			•	
TECRL	Catecholaminergic polymorphic ventricular tachycardia (CPVT)	AR	Worldwide	1 in 1661	99	1 in 166140	•	•		•	•
TF	Atransferrinemia	AR	Worldwide	1 in 398	99	1 in 39800	•			•	
TFR2	Hemochromatosis	AR	Worldwide	1 in 1047	99	1 in 104700	•			•	
TG	Thyroid dyshormonogenesis	AR	Worldwide	1 in 360	97	1 in 12000	•			•	
TGM1	Ichthyosis, congenital	AR	Worldwide	1 in 315	99	1 in 31500	•			•	
TH	Segawa syndrome	AR	Worldwide	1 in 1688	99	1 in 168800	•			•	
TMEM107	Joubert syndrome	AR	Worldwide	1 in 1800	99	1 in 180000	•			•	
TMEM216	Joubert syndrome and Meckel syndrome	AR	Worldwide	1 in 1843	99	1 in 184300	•	•	•	•	•
TMEM231	Joubert syndrome and Meckel syndrome	AR	Worldwide	1 in 831	89	1 in 7552	•			•	
TMEM67	Joubert syndrome, Meckel syndrome, Nephronophthisis and COACH syndrome	AR	Worldwide	1 in 288	96	1 in 7200	•			•	
TMPRSS3	Deafness	AR	Worldwide	1 in 122	95	1 in 2448	•			•	
TPO	Thyroid dyshormonogenesis	AR	Worldwide	1 in 293	99	1 in 29340	•			•	
TPP1	Neuronal ceroid lipofuscinosis type 2	AR	Worldwide	1 in 516	99	1 in 51600	•	•		•	•
TRAF3IP1	Senior-Loken syndrome 9	AR	Worldwide	1 in 1186	99	1 in 118620	•			•	
TREX1	Aicardi-Goutières syndrome	AR	Worldwide	1 in 831	99	1 in 83070	•			•	
TRIM37	Mulibrey nanism	AR	Worldwide	1 in 360	99	1 in 36000	•			•	
TSEN2 Excluded from the analysis: NM_001321278, exon 12	Pontocerebellar hypoplasia	AR	Worldwide	1 in 6353	87	1 in 48870	•			•	
TSEN34	Pontocerebellar hypoplasia	AR	Worldwide	1 in 13500	99	1 in 1350000	•			•	
TSEN54	Pontocerebellar hypoplasia	AR	Worldwide	1 in 416	95	1 in 8316	•			•	

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TSFM Excluded from the analysis: NM_001172696, exon 5	Combined oxidative phosphorylation deficiency	AR	Worldwide	1 in 772	92	1 in 9650	•			•	
TTC37	Trichohepatoenteric syndrome	AR	Worldwide	1 in 720	99	1 in 72000	•			•	
TTC8	Bardet-Biedl syndrome and Retinitis pigmentosa	AR	Worldwide	1 in 3600	99	1 in 360000	•			•	
TTPA	Ataxia with isolated vitamin E deficiency	AR	Worldwide	1 in 869	99	1 in 86900	•			•	
TWNK	Mitochondrial DNA depletion syndrome and Perrault syndrome	AR	Worldwide	1 in 831	99	1 in 83069	•			•	
TYMP	Mitochondrial DNA depletion syndrome	AR	Worldwide	1 in 1286	99	1 in 128600	•			•	
TYR#	Oculocutaneous albinism	AR	Worldwide	1 in 90	98	1 in 4500	•	•		•	•
UBR1	Johanson-Blizzard syndrome	AR	Worldwide	1 in 5400	99	1 in 540000	•			•	
UNC13D	Familial hemophagocytic lymphohistiocytosis	AR	Worldwide	1 in 600	90	1 in 6003	•			•	
USH1C	Deafness|Usher syndrome type IC	AR	Worldwide	1 in 981	97	1 in 32700	•			•	
USH2A	Retinitis pigmentosa 39 and Usher syndrome type 2A	AR	Worldwide	1 in 127	99	1 in 12700	•	•		•	•
VPS13A	Choreoacanthocytosis	AR	Worldwide	1 in 812	97	1 in 27067	•		•	•	
VPS13B	Cohen syndrome	AR	Worldwide	1 in 1006	99	1 in 100600	•			•	
VPS45 Excluded from the analysis: NM_001279353, exon 13	Severe congenital neutropenia type 5	AR	Worldwide	1 in 4227	97	1 in 140900	•			•	
VPS53	Pontocerebellar hypoplasia type 2E	AR	Worldwide	1 in 3869	99	1 in 386900	•			•	
VRK1	Pontocerebellar hypoplasia	AR	Worldwide	1 in 4215	99	1 in 421500	•		•	•	
XPA	Xeroderma pigmentosum	AR	Worldwide	1 in 4687	99	1 in 468700	•			•	
XPC	Xeroderma pigmentosum	AR	Worldwide	1 in 952	99	1 in 95200	•	•		•	•
ZFYVE26	Spastic paraplegia 15	AR	Worldwide	1 in 854	99	1 in 85400	•			•	
ZNF469	Brittle cornea syndrome	AR	Worldwide	1 in 3085	99	1 in 308520	•			•	

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ZNHIT3 Excluded from the analysis: ZNHIT3 NM_001281432, exon 5	PEHO syndrome	AR	Worldwide	1 in 771	77	1 in 3353	•			•	

Some genes in the Comprehensive Reproductive Screen have exons that are excluded from our target regions due to suboptimal coverage:

ADAMTS2 (NM_021599, exon 11)
CC2D2A (NM_020785, exon 7)
CHM (NM_001145414, exon 5)
KIAA0586 (NM_001244189, exons 6 and 33)
MCPH1 (NM_001322042, exon 14)
PCCB (NM_001178014, exon 4)
RPGRIP1L (NM_015272, exon 23)
TSEN2 (NM_001321278, exon 12)
TSFM (NM_001172696, exon 5)
VPS45 (NM_001279353, exon 13)
ZNHIT3 (NM_001281432, exon 5)

Special reporting policies:
CFTR: The 5T and associated TG repeat variants are not reported as their relevance in relation to classical cystic fibrosis is unclear and this test is not intended to screen for risk of congenital bilateral absence of the vas deferens (CBAVD) or CFTR-related pancreatitis risk.

For the CYP21A2 gene we report only specific variants listed below:

c.955C>T	p.(Gln319*)
c.844G>T	p.(Val282Leu)
c.293-13C>G	
c.1360C>T	p.(Pro454Ser)
c.518T>A	p.(Ile173Asn)
c.1447C>T	p.(Pro483Ser)
c.92C>T	p.(Pro31Leu)

TYR: The c.1205G>A, p.(Arg402Gln) (NM_000372.5) hypomorphic variant is typically associated with mild skin/hair/eye pigmentation changes and is therefore not reported as this test is intended to identify variants that cause severe TYR-related oculocutaneous albinism.

NGS doesn't usually detect inversions, gene conversions, balanced translocations, repeat expansions or, non-coding variants deeper than +/- 20 bps from exon-intron boundary unless otherwise indicated.

#Some, or all, of the gene is duplicated in the genome.

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