CGT Personalized v5.4.26

Patient Information		Sample Information			Clinic Information	
Unique pat id.:	1011600	**Sample type:**	Saliva		**Clinic:**	GENESIS IVF &
Patient name:	[redacted]	**Date of draw:**	01/11/2024			WOMEN'S SPECIALIST
[redacted]	[redacted]	**Date of receipt:**	27/11/2024			CENTRE
Ethnic group:	Asian	**Report date/time:**	29/12/2024	20:32	**Doctor:**	NG PENG WAH

Indication: No family history

TEST RESULTS

POSITIVE

The individual is carrier of:

Familial Mediterranean fever

Gene :	MEFV	**Allele:**	Het
DNA Change:	NM_000243.3:c.1108dupC	**Inheritance:**	AR
Protein change:	p.Gln370fs	**OMIM phenotype:**	249100
Variant classification:	Likely Pathogenic		

INTERPRETATION OF TEST RESULTS

Typically, a positive result does not have direct clinical consequences for the carrier individual. There is another normal gene copy for all positive autosomal recessive (AR) genes indicated in the table which provides normal biological information. The likelihood of transmission of the variant(s) to offspring is 50%, independent for each variant. If the partner, or gamete donor, screens negative for the pathogenic or likely pathogenic variants in the gene(s) included in the table for this patient, the reproductive risk would be reduced. Please note that family members may also carry the variant(s) reported here, and this information may be significant for them and their offspring.

If a patient and partner, or gamete donor, are both carriers of variants in the same gene associated with AR inheritance, there is a 25% chance that any child they have together would be affected. If a female patient is a carrier for an X-linked condition, there is a 50% chance that each of the reproductive couple's children would also be a carrier. Males would typically express symptoms of the condition, and females are typically unaffected or may display milder symptoms.

For genes with a negative test result, the risk of having children affected by the associated disorders decreases significantly compared to the general population. This also the case for a negative personal result when a reproductive partner or a gamete donor is a carrier for a pathogenic or likely pathogenic variant in one or more of the tested genes. However, due to test limitations associated with any genetic test, this low risk is not zero (see limitations section and informed consent form)

TEST DESCRIPTION

The Carrier Genetic Test (CGT) is a preconception DNA screening test that aims to identify individuals and couples at increased risk of conceiving children affected by a monogenic disease. Knowledge of this risk may influence a couple's decision to conceive or encourage the couple to adopt preventive measures, including preimplantation genetic testing for the at risk disease (PGT-M) prenatal genetic testing, or to use donated gametes. The multigene CGT interrogates thousands of DNA variants using a high-throughput technology (Next Generation Sequencing, NGS).

COMMENTS

None

TEST METHODOLOGY

DNA is isolated from the sample, usually blood or saliva, and analyzed by whole exome sequencing by NGS., including capture and sequence of all human exons and other gene regions of interest where known disease-causing variants are located. Sequencing raw data is then analyzed using bioinformatics (bioinformatic pipeline v3.0). Briefly, sequence alignment against the GRCh37 human genome reference, variant calling, annotation, and real-time interpretation of variants. QC parameters include, among other, that all reported samples will have a minimum of 7Gb of data, with minimal mean coverage greater than 75x, and a specific depth analysis for more than 68,000 DNA positions where known pathogenic variants are located. In addition, complementary tests (non-NGS techniques) were performed for the following genes, if included, SMN1 gene exon 7-deletion; CYP21A2 gene frequent mutations; HBA1 and HBA2 genes frequent deletions; FXN gene GAA repeat sizing; FMR1 gene CGG repeat sizing (females only); DMD gene frequent deletions/duplications; F8 gene intron 22 inversion (females only). Based on our validations studies, reported samples will have analytical detection rate for SNV variants as per the control sample NA12878 (Control positive); PASS value: NA12878 Sensitivity SNV ≥ 0.97000.

TEST LIMITATIONS

In the general population, there is a 3-5% risk for birth defects caused by genetic and/or non-genetic factors not detected by this type of test.

Analytically, the CGT test does not cover all known monogenic diseases nor all disease-causing variants for each tested gene. The test does not include the analysis of conditions associated with mitochondrial DNA nor multifactorial nor digenic inheritance. The test does not detect large rearrangements (inversions, deletions and duplications more than 15 nucleotides), variants located in regulatory regions or intronic regions outside the +/-3bp cut off (except if otherwise indicated), or in low sequence coverage areas (<7x). DNA changes caused by trinucleotide repeat expansions are not detected, except those indicated in the methodology section. For copy number variation analysis, when a normal result is obtained (2 copies detected), it is not possible to confirm that one copy is present in each of the two alleles (non-carrier) or if both copies are present in cis on the same allele, with no copies in the other allele (silent carrier). Clinical sensitivity varies among conditions. In particular, the sensitivity for SMN1 is approximately 96% because it is not possible to identify silent carriers among patients with 2 SMN1 copies detected and because point mutations or small indels are not analyzed. For the HEXB gene, the common 16 kb deletion that causes disease in 30% of affected patients is not included in CGT analysis.

Then, a negative CGT result significantly reduces but does not completely exclude the possibility of being a carrier of a variant associated with single gene disorders (see residual risk table). The presence of pseudogenes and/or rare polymorphisms and/or homopolymers may lead to false negative or false positive results. In addition, a negative result for the CGT variants does not exclude the possibility of a de novo variant occurring in the offspring. Germline mosaicism or low-level somatic mosaicism cannot be detected. As with any laboratory test, there is a small chance that this result may be inaccurate for a procedural reason such as an error during sample collection, labelling, processing, data collection or interpretation. Please note that the clinical classification of variants can change over time. To check whether there have been any changes to the classification of reported variants, please contact IGENOMIX.

LEGAL/QUALITY

This test was developed, and its performance characteristics determined by Igenomix Group. It has not been cleared or approved by the US Food and Drug Administration. The test is used as a laboratory developed test for clinical purposes.

EXEMPTION CLAUSE OF DIAGNOSTIC LIABILITY

The genetic diagnosis services carried out by IGENOMIX SPAIN LAB, SLU are exclusively intended to be interpreted by qualified/certified health professionals.

The result obtained by this test and the information that could be derived from it, cannot be considered in any case as substitute of genetic counselling or medical treatment by a trained professional neither represent itself a medical enquiry. We recommend that you consult your physician for genetic testing & counselling upon reception of your results.

Any result should be interpreted in the context of all available clinical findings, within the general context of a medical investigation, which must be conducted by clinically trained professionals. IGENOMIX SPAIN LAB, SLU is not responsible for any decisions made or actions undertaken by the contracting party based on the results provided by IGENOMIX SPAIN LAB, SLU or otherwise., nor the harmful temporary consequences diverted by its use, making specific discretion of taking appropriate legal measures assuming an improper use of those mentioned studies and analysis.

SIGNED	COUNTERSIGNED
Ana Bover	**María Lairón PhD**
3068-CV	3647-CV
Testing Personnel	Testing Personnel

Lab CLIA No.: 99D2146167

Igenomix®
PART OF VITROLIFE GROUP®

Familial Mediterranean fever

What is Familial Mediterranean fever?

Familial Mediterranean fever follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MEFV gene located on chromosomal region 16p13.3. The age of onset is infantile or adult (before the age of 30). This disease is characterized by recurrent short episodes of fever and serositis resulting in pain in the abdomen, chest, joints and muscles. In rare cases, this condition appears to be inherited in an autosomal dominant pattern. The prevalence is 1:10,000-5:10,000.

What is the next step if I am a carrier of Familial Mediterranean fever?

If you are a carrier of Familial Mediterranean fever it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

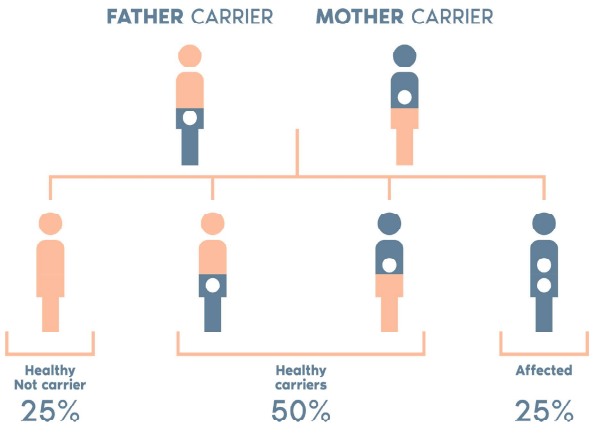
If your partner tests negative for Familial Mediterranean fever, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Familial Mediterranean fever?

When both parents are carriers of Familial Mediterranean fever, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

In this case it is advisable to use the same assay (CGT) to test candidate donors and choose one that is negative for the same condition.



If both are carriers of the disease contact your doctor or genetic counselor for information on genetic options for family planning.

LIST OF ANALYZED GENES

Total genes analyzed: 500. Genes analyzed AR: 468. Genes analyzed XL: 32.

Gene mean coverage >100x ALDOA, NAGA, AGT, AGTR1, ACE, DDC, COL17A1, CDH3, CAPN3, TYRP1, CTSD, HSPD1, CLCN1, COL4A3, COL7A1, COL4A4, COL1A2, COL9A1, COL11A1, COL18A1, C3, GJB2, GJA1, CNGA1, DSP, ERCC2, ETFB, EDN3, EDNRB, ERCC3, ERCC5, CFH, FGA, FGB, FH, MPV17, HBA1, HBA2, HBB, HGF, HSPG2, HADHB, IGF1, ITGA6, ITGB4, INSR, LDHA, LAMC2, LAMB3, LIFR, PLOD1, MAK, LAMA2, MTTP, MYO5A, NEB, NEFL, PEX2, ALPL, PGM1, PHKG2, POU1F1, ENPP1, PLG, POLG, PSAP, RDX, RAG1, RAG2, REN, RPE65, PDE6A, PDE6B, RLBP1, RHO, RYR1, SAG, STIL, TERT, TK2, TSHB, TTN, TNNT1, TH, NTRK1, DPAGT1, UGT1A1, ETFDH, PCCA, PCCB, GLDC, AMT, DLD, DBT, BCKDHB, MVK, F11, MYO7A, ATP7A, DKC1, ARSL, BTK, DMD, EMD, CD40LG, WAS, MTM1, OTC, SH2D1A, PDHA1, OCRL, F9, RP2, GPR143, F8, HPRT1, IL2RG, L1CAM, FMR1, PRPS1, RPGR, LBR, LRP2, SGCA, JAK3, GUCY2D, SCNN1A, ACADSB, KCNJ1, PEX5, TTPA, IGHMBP2, RELN, AUH, STAR, CPT2, PPT1, CNGB1, SCNN1B, SCNN1G, LAMA3, DDB2, PDE6C, SLC12A1, HADHA, SGCB, MYO6, DMP1, GSS, NPC2, CTSK, CSTB, ATR, PLEC, DGUOK, PROP1, RAPSN, ABCA4, ATIC, PEX7, PEX12, VDR, PMM2, LIG4, HSD17B4, PHYH, PEX1, HTRA1, TULP1, SC5D, TRIM32, CFTR, RAB3GAP1, TECTA, MGAT2, MYO15A, SLC37A4, NPHS1, CLCN7, DLL3, SPG7, ADGRV1, DHCR7, DYSF, AGPS, KCNJ13, PNPO, GLE1, TSHR, PRKRA, ASS1, DPM1, LRP5, MTMR2, LARGE1, PLA2G6, SLC24A1, BCS1L, OTOF, MOCS1, MOCS2, ELP1, GNE, SLC25A13, SLC25A15, RAB27A, EIF2AK3, SNAP29, CRB1, CRLF1, GRHPR, SLC17A5, PROM1, AIPL1, GJB6, NR2E3, TCAP, SACS, IDH3B, ADAMTS2, ALG6, TCIRG1, MERTK, TFR2, TSFM, LRAT, SLC12A6, TBCE, SLC26A5, HPS1, TRIM37, CNGB3, USH1C, MCOLN1, NDRG1, SGSH, ASPM, CRTAP, TMPRSS3, PCDH15, CDH23, SLC35A1, SLC26A4, MRPS22, MLC1, VPS13A, UBR1, PDP1, TWNK, RAB23, BSCL2, WFS1, SELENON, MFRP, WNT10A, CTNS, APTX, CLRN1, NOP10, ZMPSTE24, GDAP1, TREX1, GFM1, PKHD1, TMC1, SLC26A2, CLN6, GAA, MYO3A, ALDH4A1, POMGNT1, ALMS1, HEXB, ATP7B, ACADS, SUOX, GALC, TYR, COQ8A, GALT, ACADM, IVD, OTOA, IMPG2, NPHP1, SPART, FANCA, NPHP4, SEMA4A, POMT1, PDSS1, POMT2, FKTN, SBDS, HGD, MMAA, LRPPRC, MMAB, KCNV2, SMPD1, NPC1, OSTM1, GNS, USH1G, SBF2, ACAT1, FRAS1, GBE1, GNPTAB, BEST1, WHRN, TPP1, NPHP3, SIL1, ETFA, MEFV, DHDDS, CDK5RAP2, SH3TC2, NAGS, CPS1, ASL, ARG1, BCKDHA, CERKL, USH2A, PLCE1, MCEE, ETHE1, PYGM, SETX, VPS33B, CYP4V2, ANO5, TSEN54, PC, GCDH, RDH12, AHI1, SGCG, FREM2, ADA, DNAJC19, MCCC1, MCCC2, MMUT, FLVCR1, D2HGDH, IQCB1, RAB3GAP2, CENPJ, ESCO2, FIG4, LHFPL5, MAN2B1, CDHR1, ACADVL, FANCM, HPD, PKLR, ACOX1, TRIOBP, COQ2, MKS1, TMEM67, ALDH5A1, CEP290, SLC4A11, PJVK, P3H1, HGSNAT, MAT1A, HIBCH, STRA6, SPG11, AGL, PSAT1, RPGRIP1L, G6PC3, PLEKHG5, ACAD9, FGD4, MFSD8, XPA, IFT80, SUCLG1, KIF7, OCA2, GLB1, CA2, GUSB, ARSB, MMADHC, ZFYVE26, CC2D2A, UQCRQ, FUCA1, PAH, LRTOMT, EYS, ALDOB, DPYD, PDZD7, TAT, INPP5E, LOXHD1, GM2A, AGA, TMEM216, GRXCR1, OAT, CBS, CEP152, WDR62, G6PC1, CYP21A2, FAH, HMGCL, FANCC, GPR179

Gene mean coverage 50x-100x B4GALT1, MPI, PDE6G, VLDLR, SLC6A8, ARX, PLP1, NR0B1, CPT1A, LHX3, FOXN1, GAMT, HESX1, RAX, ESRRB, CYP7B1, GRM6, AGXT, MKKS, CHST6, CLDN14, PRX, NUP62, SLC35C1, ALG1, DCLRE1C, SLC45A2, SIX6, ESPN, STRC, NHP2, HEXA, CLN3, MCPH1, TMIE, ARSA, ASPA, CLN5, GJC2, ARL6, ARL13B, MRPS16, SLC25A22, ERCC8, ERCC6, MMACHC, CLDN19, MED25, PDSS2, MARVELD2, PRCD, HYLS1, FAM20C, ISCU, B9D2, ZNF469, ADAMTSL2, TPRN

Gene mean coverage < 50x MECP2, POU3F4, EDA, IDS, AR, SMN1, PDX1, WNT7A, NEUROG3, GAN, PANK2, BSND, FKRP, MLYCD, FXN, CLN8, PEX26, NMNAT1, BTD, SLC35D1

GLOSSARY

TYPES OF INHERITANCE:

- AR: Autosomal recessive

  Inherited conditions that require two pathogenic variants (one from each parent) in a given gene to display symptoms.
- XR: X-linked recessive

  The gene is located on the X chromosome. Men with a pathogenic variant have the disease. Women with a pathogenic variant are carriers and generally asymptomatic or may mild symptoms.
- Digenic inheritance

  In some diseases, the symptoms could be explained by the coexistence of pathogenic variants in two different genes related with the disease instead of two pathogenic variants in the same gene.

ALLELES:

Pathogenic variants present in the two copies of a gene.

- Homozygous pathogenic variant (Hom.):

  Each copy of the gene has the same pathogenic variant. Generally, this is associated with clinical symptoms.
- Compound heterozygous (Het.):

  Each copy of the gene has a different pathogenic variant. Generally, this is associated with clinical symptoms. This situation is referred as having variants "in trans".

Pathogenic variant present in one copy of a gene.

- Heterozygous pathogenic variant (Het.):
  Only one copy of a gene has a pathogenic variant. There is another normal gene copy.

Note: Sometimes an individual has two pathogenic variants in the same gene copy. This situation is referred as having variants in cis and it is considered as a single pathogenic variant.

CNV:

Refers to copy number variation (deletion or duplication), i.e., the number of copies of a particular gene (or gene region) is different from the usual two copies.

LARGE GENE CONVERSION:

Refers to pathogenic variants caused by gene sequence exchange or replacement between a normal functional gene and a quasi-identical non-functional gene (pseudogene).

X-linked conditions				
Chrom	**Gene**	**Disease/Condition**	**Carrier Rate**	**Residual Risk**
X	AR	Androgen insensitivity syndrome	1 in 6250	1 in 10417
X	ARSL	Chondrodysplasia punctata, brachytelephalangic	< 1 in 100 000	Reduced
X	ARX	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	1 in 25 000	1 in 100000
X	ATP7A	Menkes disease; Occipital horn syndrome	1 in 25000	1 in 100000
X	BTK	Agammaglobulinemia X-linked, type 1	1 in 50,000	1 in 333333
X	CD40LG	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	< 1 in 100 000	Reduced
X	DKC1	Dyskeratosis congenita, X-linked	1 in 62500	1 in 1250000
X	DMD	DMD-related conditions	1 in 1374	1 in 27480
X	EDA	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	1 in 2500	1 in 16667
X	EMD	Emery-Dreifuss muscular dystrophy, type 1, X-linked	< 1 in 100 000	Reduced
X	F8	Hemophilia A	1 in 1250	1 in 31250
X	F9	Hemophilia B	1 in 6250	1 in 62500
X	FMR1	FMR1-related conditions	1 in 400	1 in 40000
X	GPR143	Ocular albinism, type 1 (Nettleship-Falls type)	1 in 15000	1 in 18750
X	HPRT1	Lesch-Nyhan syndrome	1 in 95000	1 in 380000
X	IDS	Mucopolysaccharidosis, type 2	1 in 25000	1 in 125000
X	IL2RG	Severe combined immunodeficiency, X-linked	1 in 25000	1 in 500000
X	L1CAM	L1 Syndrome	1 in 7500	1 in 150000
X	MECP2	Encephalopathy, neonatal severe; Rett syndrome	1 in 37500	1 in 250000
X	MTM1	Myotubular myopathy, X-linked	1 in 12500	1 in 83333
X	NR0B1	Adrenal hypoplasia, congenital	1 in 17500	1 in 58333
X	OCRL	Lowe Syndrome; Dent disease type 2	< 1 in 100 000	Reduced
X	OTC	Ornithine transcarbamylase deficiency	1 in 50000	1 in 166667
X	PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	< 1 in 100 000	Reduced
X	PLP1	Pelizaeus-Merzbacher disease	1 in 353	1 in 441
X	POU3F4	Deafness, X-linked, type 2	1 in 556,112	<1 in 1,000,000
X	PRPS1	PRPS1-related disoders	< 1 in 100 000	Reduced
X	RP2	Retinitis pigmentosa, type 2, X-linked	1 in 5000	1 in 62500
X	RPGR	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	1 in 20000	1 in 28571
X	SH2D1A	Lymphoproliferative syndrome, X-linked, type 1	< 1 in 100 000	Reduced
X	SLC6A8	Cerebral creatine deficiency syndrome, type 1	< 1 in 100 000	Reduced
X	WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	< 1 in 100 000	Reduced

Autosomal recessive conditions				
Chrom	**Gene**	**Disease/Condition**	**Carrier Rate**	**Residual Risk**
1	ABCA4	Stargardt disease 1; Retinitis pigmentosa 19; Cone-rod dystrophy 3	1 in 62	1 in 119
3	ACAD9	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	1 in 309	1 in 3090
1	ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	1 in 60	1 in 600
12	ACADS	Short-chain acyl-CoA dehydrogenase deficiency	1 in 94	1 in 1880
10	ACADSB	Short/branched-chain acyl-CoA dehydrogenase deficiency	1 in 500	1 in 1,125
17	ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	1 in 112	1 in 1120
11	ACAT1	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	1 in 300	1 in 3750
17	ACE	Renal tubular dysgenesis	< 1 in 500	Reduced
17	ACOX1	Peroxisomal acyl-CoA oxidase deficiency	< 1 in 500	Reduced
20	ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	1 in 390	1 in 2600
5	ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	< 1 in 500	Reduced
9	ADAMTSL2	Geleophysic dysplasia type 1	< 1 in 500	Reduced
5	ADGRV1	Usher syndrome, type 2C	1 in 80	1 in 147
4	AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)	< 1 in 500	Reduced
1	AGL	Glycogen storage disease, type 3	1 in 200	1 in 2000
2	AGPS	Rhizomelic chondrodysplasia punctata, type 3	< 1 in 500	Reduced
1	AGT	Renal tubular dysgenesis	< 1 in 500	Reduced
3	AGTR1	Renal tubular dysgenesis	< 1 in 500	Reduced
2	AGXT	Hyperoxaluria, primary, type 1	1 in 174	1 in 2486
6	AHI1	Joubert syndrome, type 3	1 in 334	1 in 706
17	AIPL1	Leber congenital amaurosis, type 4	1 in 400	1 in 571
1	ALDH4A1	Hyperprolinemia, type 2	1 in 500	1 in 49,951
6	ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	N/A	N/A
16	ALDOA	Glycogen storage disease type 12	< 1 in 500	Reduced
9	ALDOB	Fructose intolerance, hereditary	1 in 80	1 in 400
16	ALG1	Congenital disorder of glycosylation, type 1K	1 in 87	1 in 130
1	ALG6	Congenital disorder of glycosylation, type 1C	1 in 500	1 in 5000
2	ALMS1	Alström syndrome	1 in 250	1 in 1667
1	ALPL	ALPL-related conditions	1 in 274	1 in 2740
3	AMT	Glycine encephalopathy	1 in 310	1 in 6200
11	ANO5	Limb-girdle muscular dystrophy, type 12 (LGMD R12)	< 1 in 500	Reduced
9	APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	< 1 in 500	Reduced
6	ARG1	Argininemia (arginase deficiency)	1 in 418	1 in 13933
3	ARL13B	Joubert syndrome type 8	1 in 72	1 in 119
3	ARL6	Bardet-Biedl syndrome, type 3	< 1 in 500	Reduced
22	ARSA	Metachromatic leukodystrophy	1 in 192	1 in 1920
5	ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	1 in 314	1 in 3925
7	ASL	Argininosuccinic aciduria	1 in 133	1 in 665
17	ASPA	Canavan disease	1 in 416	1 in 13867
1	ASPM	Primary microcephaly type 5, autosomal recessive	< 1 in 500	Reduced
9	ASS1	Citrullinemia, type 1	1 in 300	1 in 3750
2	ATIC	AICA-ribosiduria due to ATIC deficiency	< 1 in 500	Reduced
13	ATP7B	Wilson disease	1 in 90	1 in 450
3	ATR	Seckel syndrome, type 1	< 1 in 500	Reduced
9	AUH	3-methylglutaconic aciduria, type 1	< 1 in 500	<1 in 938
9	B4GALT1	Congenital disorder of glycosylation, type 2D	< 1 in 500	<1 in 50,000
19	B9D2	Joubert syndrome, type 34; ?Meckel syndrome, type 10	< 1 in 500	Reduced
19	BCKDHA	Maple syrup urine disease, type 1A	1 in 320	1 in 3200
6	BCKDHB	Maple syrup urine disease, type 1B	1 in 365	1 in 2808
2	BCS1L	Mitochondrial complex III deficiency nuclear type 1; GRACILE syndrome; Bjornstad syndrome	1 in 320	1 in 2133
11	BEST1	Bestrophinopathy, AR	< 1 in 500	Reduced
11	BSCL2	Congenital generalized lipodystrophy, type 2; Encephalopathy, progressive, with or without lipodystrophy	< 1 in 500	Reduced
1	BSND	Bartter syndrome, type 4A	< 1 in 500	Reduced
3	BTD	Biotinidase deficiency	1 in 120	1 in 1500
19	C3	Complement component 3 deficiency	N/A	N/A
8	CA2	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	< 1 in 500	<1 in 1,000
15	CAPN3	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	1 in 103	1 in 412
21	CBS	Homocystinuria due to cystathionine beta-synthase	1 in 274	1 in 2740
4	CC2D2A	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2	1 in 196	1 in 2,800
10	CDH23	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D	1 in 216	1 in 1080
16	CDH3	Ectodermal dysplasia, ectrodactyly, and macular dystrophy; Hypotrichosis, congenital, with juvenile macular dystrophy	N/A	N/A
10	CDHR1	Cone-rod dystrophy, type 15	< 1 in 500	Reduced
9	CDK5RAP2	Primary microcephaly type 3, autosomal recessive	N/A	N/A
13	CENPJ	Primary microcephaly type 6, autosomal recessive	< 1 in 500	Reduced
15	CEP152	Primary microcephaly type 9, autosomal recessive	N/A	N/A

12	CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	1 in 150	1 in 375
2	CERKL	Retinitis pigmentosa, type 26	1 in 250	1 in 1667
1	CFH	Complement factor H deficiency	< 1 in 500	Reduced
7	CFTR	Cystic fibrosis	1 in 25	1 in 833
16	CHST6	Macular corneal dystrophy	1 in 80	1 in 394
7	CLCN1	Myotonia congenita, recessive	1 in 159	1 in 319
16	CLCN7	Osteopetrosis, autosomal recessive type 4	< 1 in 500	Reduced
21	CLDN14	Deafness type 29, autosomal recessive	< 1 in 500	Reduced
1	CLDN19	Rena hypomagnesemia type 5, with ocular involvement	< 1 in 500	Reduced
16	CLN3	Ceroid lipofuscinosis, neuronal, type 3	1 in 242	1 in 346
13	CLN5	Ceroid lipofuscinosis, neuronal, type 5	1 in 400	1 in 8000
15	CLN6	Ceroid lipofuscinosis, neuronal, type 6	< 1 in 500	Reduced
8	CLN8	Ceroid lipofuscinosis, neuronal, type 8	< 1 in 500	Reduced
3	CLRN1	Usher syndrome, type 3A	1 in 250	1 in 1667
4	CNGA1	Retinitis pigmentosa type 49	1 in 625	1 in 1,171
16	CNGB1	Retinitis pigmentosa type 45	1 in 200	1 in 4000
8	CNGB3	Achromatopsia, type 3	1 in 125	1 in 1250
1	COL11A1	Fibrochondrogenesis type 1	1 in 500	1 in 16666
10	COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type	< 1 in 500	Reduced
21	COL18A1	Knobloch syndrome, type 1	< 1 in 500	Reduced
7	COL1A2	Ehlers-Danlos syndrome, cardiac valvular type	N/A	N/A
2	COL4A3	Alport syndrome, autosomal recessive, type 2	1 in 300	1 in 1500
2	COL4A4	Alport syndrome, autosomal recessive, type 2	1 in 425	1 in 4250
3	COL7A1	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	1 in 150	1 in 1000
6	COL9A1	Stickler syndrome, type 4	N/A	N/A
4	COQ2	Primary coenzyme Q10 deficiency, type 1	< 1 in 500	Reduced
1	COQ8A	Primary coenzyme Q10 deficiency, type 4	< 1 in 500	Reduced
2	CPS1	Carbamoylphosphate synthetase 1 deficiency	1 in 500	1 in 2500
11	CPT1A	Carnitine palmitoyltransferase type 1A deficiency, hepatic	< 1 in 500	Reduced
1	CPT2	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	1 in 100	1 in 667
1	CRB1	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	1 in 158	1 in 316
19	CRLF1	Cold-induced sweating syndrome type 1	< 1 in 500	Reduced
3	CRTAP	Osteogenesis imperfecta, type 7	1 in 1,416	1 in 3,539
21	CSTB	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)	< 1 in 500	Reduced
17	CTNS	Nephropathic cystinosis	1 in 200	1 in 400
11	CTSD	Ceroid lipofuscinosis, neuronal, type 10	< 1 in 500	Reduced
1	CTSK	Pycnodysostosis	< 1 in 500	Reduced
6	CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	1 in 62	1 in 1240
4	CYP4V2	Bietti crystalline corneoretinal dystrophy	1 in 130	1 in 1300
8	CYP7B1	Spastic paraplegia, type 5A, autosomal recessive	< 1 in 500	Reduced
2	D2HGDH	D-2-hydroxyglutaric aciduria	< 1 in 500	Reduced
1	DBT	Maple syrup urine disease, type 2	1 in 410	1 in 2733
10	DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	< 1 in 500	Reduced
11	DDB2	Xeroderma pigmentosum, complementation group E	< 1 in 500	Reduced
7	DDC	Aromatic L-amino acid decarboxylase deficiency	N/A	N/A
2	DGUOK	DGUOK-related mitochondrial DNA depletion syndrome	< 1 in 500	Reduced
11	DHCR7	Smith-Lemli-Opitz syndrome	1 in 100	1 in 1000
1	DHDDS	Retinitis pigmentosa, type 59	< 1 in 500	Reduced
7	DLD	Dihydrolipoamide dehydrogenase deficiency	< 1 in 500	Reduced
19	DLL3	Spondylocostal dysostosis type 1	N/A	N/A
4	DMP1	Hypophosphatemic rickets, autosomal recessive	< 1 in 500	Reduced
3	DNAJC19	3-methylglutaconic aciduria, type 5	< 1 in 500	Reduced
11	DPAGT1	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	< 1 in 500	<1 in 808
20	DPM1	Congenital disorder of glycosylation, type 1E	< 1 in 500	<1 in 1,750
1	DPYD	Dihydropyrimidine dehydrogenase deficiency	1 in 558	1 in 55,701
6	DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma; Epidermolysis bullosa, lethal acantholytic	< 1 in 500	Reduced
2	DYSF	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	1 in 300	1 in 3000
20	EDN3	Waardenburg syndrome, type 4B	< 1 in 500	Reduced
13	EDNRB	ABCD syndrome	< 1 in 500	Reduced
2	EIF2AK3	Wolcott-Rallison syndrome	< 1 in 500	<1 in 2,500
9	ELP1	Familial dysautonomia	1 in 200	1 in 2000
6	ENPP1	Arterial calcification, generalized, of infancy, type 1	1 in 333	1 in 3330
19	ERCC2	Trichothiodystrophy, type 1; Xeroderma pigmentosum, group D	1 in 500	1 in 10000
2	ERCC3	Trichothiodystrophy, type 2	1 in 436	1 in 1,306
13	ERCC5	Cerebrooculofacioskeletal syndrome 3; Xeroderma pigmentosum, group G;Xeroderma pigmentosum, group G/Cockayne syndrome	< 1 in 500	Reduced
10	ERCC6	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	1 in 300	1 in 2000

5	ERCC8	Cockayne syndrome, type A	< 1 in 500	Reduced
8	ESCO2	Roberts syndrome	< 1 in 500	Reduced
1	ESPN	Deafness, autosomal recessive, type 36	N/A	N/A
14	ESRRB	Deafness, autosomal recessive, type 35	< 1 in 500	Reduced
15	ETFA	Glutaric acidemia, type 2A	< 1 in 500	Reduced
19	ETFB	Glutaric acidemia, type 2B	< 1 in 500	Reduced
4	ETFDH	Glutaric acidemia, type 2C	1 in 300	1 in 2000
19	ETHE1	Ethylmalonic encephalopathy	< 1 in 500	Reduced
6	EYS	Retinitis pigmentosa, type 25	1 in 100	1 in 1000
4	F11	Factor XI deficiency	1 in 200	1 in 2500
15	FAH	Tyrosinemia, type 1	1 in 200	1 in 2000
7	FAM20C	Raine syndrome	< 1 in 500	<1 in 1,000
16	FANCA	Fanconi anemia, complementation group A	1 in 200	1 in 400
9	FANCC	Fanconi anemia, complementation group C	1 in 480	1 in 2400
14	FANCM	Spermatogenic failure, type 28; ?Premature ovarian failure 15	< 1 in 500	Reduced
4	FGA	Afibrinogenemia, congenital	N/A	N/A
4	FGB	Congenital afibrinogenemia	< 1 in 500	Reduced
12	FGD4	Charcot-Marie-Tooth disease, type 4H	N/A	N/A
1	FH	Fumarase deficiency	1 in 500	1 in 3333
6	FIG4	Charcot-Marie-Tooth disease, type 4J; Yunis-Varon syndrome	< 1 in 500	Reduced
19	FKRP	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])	1 in 176	1 in 2514
9	FKTN	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy type 13 [LGMD R13])	< 1 in 500	Reduced
1	FLVCR1	Posterior column ataxia-retinitis pigmentosa syndrome	N/A	N/A
17	FOXN1	T-cell immunodeficiency, congenital alopecia and nail dystrophy	< 1 in 500	Reduced
4	FRAS1	Fraser syndrome, type 1	1 in 300	1 in 576
13	FREM2	Fraser syndrome, type 2	N/A	N/A
1	FUCA1	Fucosidosis	1 in 1,149	1 in 4,880
9	FXN	Friedreich ataxia	1 in 91	1 in 1,014
17	G6PC1	Glycogen storage disease, type 1A	1 in 300	1 in 3000
17	G6PC3	Dursun syndrome	< 1 in 500	<1 in 1,170
17	GAA	Glycogen storage disease, type 2	1 in 100	1 in 500
14	GALC	Krabbe disease	1 in 120	1 in 218
9	GALT	Galactosemia	1 in 109	1 in 727
19	GAMT	Cerebral creatine deficiency syndrome, type 2	1 in 500	1 in 10000
16	GAN	Giant axonal neuropathy, type 1	< 1 in 500	Reduced
1	GBA1	Gaucher Disease, type I-III; GD IIIC; GD, perinatal lethal	1 in 125	1 in 1563
3	GBE1	Glycogen storage disease, type 4	1 in 192	1 in 960
19	GCDH	Glutaricaciduria, type 1	1 in 200	1 in 4000
8	GDAP1	Charcot-Marie-Tooth disease, recessive intermediate, type A	1 in 130	1 in 298
3	GFM1	Combined oxidative phosphorylation deficiency, type 1	1 in 450	1 in 1500
6	GJA1	Craniometaphyseal dysplasia, autosomal recessive	< 1 in 500	Reduced
13	GJB2	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	1 in 40	1 in 500
13	GJB6	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	1 in 421	1 in 42,000
1	GJC2	Spastic paraplegia, type 44, autosomal recessive	< 1 in 500	Reduced
3	GLB1	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	1 in 277	1 in 2770
9	GLDC	Glycine encephalopathy	1 in 180	1 in 720
9	GLE1	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	1 in 350	1 in 3500
5	GM2A	GM2-gangliosidosis, AB variant	< 1 in 500	Reduced
9	GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)	1 in 203	1 in 4060
12	GNPTAB	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	1 in 176	1 in 17,522
12	GNS	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	< 1 in 500	Reduced
17	GPR179	Night blindness, congenital stationary (complete), type 1E, autosomal recessive	< 1 in 500	Reduced
9	GRHPR	Hyperoxaluria, primary, type 2	1 in 433	1 in 21650
5	GRM6	Night blindness, congenital stationary (complete), type 1B, autosomal recessive	< 1 in 500	Reduced
4	GRXCR1	Deafness, autosomal recessive, type 25	N/A	N/A
20	GSS	Glutathione synthetase deficiency	< 1 in 500	Reduced
17	GUCY2D	Leber congenital amaurosis, type 1	1 in 248	1 in 305
7	GUSB	Mucopolysaccharidosis, type 7	1 in 552	1 in 1,6531
2	HADHA	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	1 in 250	1 in 5000
2	HADHB	Mitochondrial trifunctional protein deficiency	< 1 in 500	Reduced
16	HBA1	Alpha thalassemia	1 in 30	1 in 200
16	HBA2	Alpha thalassemia	1 in 30	1 in 200
11	HBB	HBB-related hemoglobinopathies	1 in 67	1 in 670
3	HESX1	Growth hormone deficiency with pituitary anomalies	< 1 in 500	Reduced
15	HEXA	Tay-Sachs disease	1 in 250	1 in 1250
5	HEXB	Sandhoff disease, infantile, juvenile, and adult forms	1 in 202	1 in 1347
3	HGD	Alkaptonuria	1 in 250	1 in 2500

7	HGF	Deafness, autosomal recessive, type 39	< 1 in 500	Reduced
8	HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	1 in 345	1 in 4313
2	HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency	N/A	N/A
1	HMGCL	HMG-CoA lyase deficiency	< 1 in 500	Reduced
12	HPD	Tyrosinemia, type 3	< 1 in 500	Reduced
10	HPS1	Hermansky-Pudlak syndrome, type 1	1 in 493	1 in 4930
5	HSD17B4	D-bifunctional protein deficiency	1 in 534	1 in 13350
2	HSPD1	Leukodystrophy, hypomyelinating, type 4	< 1 in 500	Reduced
1	HSPG2	Schwartz-Jampel syndrome, type 1; Dyssegmental dysplasia, Silverman-Handmaker type	< 1 in 500	<1 in 1,625
10	HTRA1	CARASIL syndrome	N/A	N/A
7	HYCC1	Leukodystrophy, hypomyelinating, type 5	< 1 in 500	Reduced
11	HYLS1	Hydrolethalus syndrome	1 in 500	1 in 714
20	IDH3B	Retinitis pigmentosa, type 46	1 in 500	1 in 999
3	IFT80	Short-rib thoracic dysplasia, type 2, with or without polydactyly	N/A	N/A
12	IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency	< 1 in 500	Reduced
11	IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S	< 1 in 500	<1 in 4,000
3	IMPG2	Retinitis pigmentosa, type 56	N/A	N/A
9	INPP5E	Joubert syndrome, type 1	< 1 in 500	Reduced
19	INSR	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A	< 1 in 500	Reduced
3	IQCB1	Senior-Loken syndrome, type 5	N/A	N/A
12	ISCU	Myopathy with lactic acidosis, hereditary	< 1 in 500	Reduced
2	ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis	N/A	N/A
17	ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia	< 1 in 500	Reduced
15	IVD	Isovaleric acidemia	1 in 115	1 in 1917
19	JAK3	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	1 in 475	1 in 732
11	KCNJ1	Bartter syndrome, type 2	< 1 in 500	Reduced
2	KCNJ13	Leber congenital amaurosis, type 16	< 1 in 500	Reduced
9	KCNV2	Retinal cone dystrophy, type 3B	< 1 in 500	Reduced
15	KIF7	Acrocallosal syndrome; Joubert syndrome, type 12	N/A	N/A
6	LAMA2	LAMA2-related muscular dystrophy	1 in 125	1 in 625
18	LAMA3	Epidermolysis bullosa, junctional 2A, intermediate;Epidermolysis bullosa, junctional 2B, severe;Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous	< 1 in 500	Reduced
1	LAMB3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	1 in 222	1 in 11100
1	LAMC2	Epidermolysis bullosa, junctional 3A, intermediate;Epidermolysis bullosa, junctional 3B, severe	< 1 in 500	Reduced
22	LARGE1	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	1 in 123	1 in 287
1	LBR	Greenberg skeletal dysplasia	N/A	N/A
11	LDHA	Glycogen storage disease type 11	< 1 in 500	Reduced
6	LHFPL5	Deafness, autosomal recessive, type 67	< 1 in 500	Reduced
9	LHX3	Pituitary hormone deficiency, combined, type 3	1 in 1,398	1 in 13980
5	LIFR	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	< 1 in 500	Reduced
13	LIG4	LIG4 syndrome	N/A	N/A
18	LOXHD1	Deafness, autosomal recessive, type 77	1 in 150	1 in 1500
4	LRAT	Leber congenital amaurosis type 14	< 1 in 500	Reduced
2	LRP2	Donnai-Barrow syndrome	< 1 in 500	Reduced
11	LRP5	Osteoporosis-pseudoglioma syndrome	< 1 in 500	Reduced
2	LRPPRC	Leigh syndrome, French-Canadian type	< 1 in 500	Reduced
11	LRTOMT	Deafness, autosomal recessive, type 63	< 1 in 500	Reduced
6	MAK	Retinitis pigmentosa type 62	N/A	N/A
19	MAN2B1	Alpha-mannosidosis	1 in 274	1 in 5480
5	MARVELD2	Deafness, autosomal recessive, type 49	N/A	N/A
10	MAT1A	Methionine adenosyltransferase deficiency, autosomal recessive	< 1 in 500	Reduced
3	MCCC1	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	1 in 353	1 in 7060
5	MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	1 in 204	1 in 4080
2	MCEE	Methylmalonyl-CoA epimerase deficiency	1 in 500	1 in 50,000
19	MCOLN1	Mucolipidosis type 4	1 in 1,166	1 in 4,850
8	MCPH1	Microcephaly type 1, primary, autosomal recessive	1 in 500	1 in 8,333
19	MED25	Basel-Vanagait-Smirin-Yosef syndrome	< 1 in 500	Reduced
16	MEFV	Familial Mediterranean fever	1 in 40	1 in 133
2	MERTK	Retinitis pigmentosa type 38	1 in 500	1 in 2500
11	MFRP	Microphthalmia, isolated type 5	1 in 250	1 in 1667
4	MFSD8	Ceroid lipofuscinosis, neuronal, type 7	1 in 300	1 in 3000
14	MGAT2	Congenital disorder of glycosylation, type 2a	< 1 in 500	Reduced
20	MKKS	Bardet-Biedl syndrome type 6	< 1 in 500	Reduced
17	MKS1	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	1 in 246	1 in 2460
22	MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	< 1 in 500	Reduced
16	MLYCD	Malonyl-CoA decarboxylase deficiency	1 in 500	1 in 1,000
4	MMAA	Methylmalonic aciduria, vitamin B12-responsive	1 in 677	1 in 4513
12	MMAB	Methylmalonic aciduria, vitamin B12-responsive, type cblB	< 1 in 500	Reduced

1	MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	1 in 170	1 in 2429
2	MMADHC	Homocystinuria, cblD type, variant 1	< 1 in 500	Reduced
6	MMUT	Methylmalonic aciduria, mut(0) type	1 in 135	1 in 3375
6	MOCS1	Molybdenum cofactor deficiency A	1 in 350	1 in 3500
5	MOCS2	Molybdenum cofactor deficiency B	1 in 400	1 in 4000
15	MPI	Congenital disorder of glycosylation, type 1B	1 in 473	1 in 11825
2	MPV17	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	1 in 612	1 in 7650
10	MRPS16	Combined oxidative phosphorylation deficiency 2	< 1 in 500	Reduced
3	MRPS22	Combined oxidative phosphorylation deficiency type 5	N/A	N/A
11	MTMR2	Charcot-Marie-Tooth disease, type 4B1	1 in 500	1 in 1,000
4	MTTP	Abetalipoproteinemia	< 1 in 500	Reduced
12	MVK	Mevalonic aciduria	1 in 286	1 in 2,261
17	MYO15A	Deafness, autosomal recessive, type 3	N/A	N/A
10	MYO3A	Deafness, autosomal recessive, type 30	< 1 in 500	Reduced
15	MYO5A	Griscelli syndrome, type 1	N/A	N/A
6	MYO6	Deafness, autosomal recessive, type 37	< 1 in 500	Reduced
11	MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	1 in 129	1 in 2580
22	NAGA	Schindler disease, type I; Schindler disease, type III; Kanzaki disease	1 in 500	1 in 5000
17	NAGS	N-acetylglutamate synthase deficiency	< 1 in 500	Reduced
8	NDRG1	Charcot-Marie-Tooth disease, type 4D	< 1 in 500	Reduced
2	NEB	Nemaline myopathy type 2	1 in 175	1 in 2188
8	NEFL	Charcot-Marie-Tooth disease, type 1F	< 1 in 500	Reduced
10	NEUROG3	Diarrhea 4, malabsorptive, congenital	< 1 in 500	Reduced
5	NHP2	Dyskeratosis congenita, autosomal recessive type 2	1 in 250	1 in 24,964
1	NMNAT1	Leber congenital amaurosis 9; Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis	< 1 in 500	Reduced
15	NOP10	Dyskeratosis congenita, autosomal recessive type 1	1 in 250	1 in 500
18	NPC1	Niemann-Pick disease, type C1	1 in 163	1 in 652
14	NPC2	Niemann-pick disease, type C2	< 1 in 500	Reduced
2	NPHP1	Joubert syndrome type 4	1 in 418	1 in 825
3	NPHP3	Meckel syndrome type 7	< 1 in 500	Reduced
1	NPHP4	Nephronophthisis type 4	< 1 in 500	Reduced
19	NPHS1	Nephrotic syndrome, type 1	1 in 112	1 in 1400
15	NR2E3	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	1 in 278	1 in 5560
1	NTRK1	Insensitivity to pain, congenital, with anhidrosis	1 in 1,122	1 in 11220
19	NUP62	Striatonigral degeneration, infantile	< 1 in 500	Reduced
10	OAT	Gyrate atrophy of choroid and retina	< 1 in 500	Reduced
15	OCA2	Oculocutaneous albinism type 2	1 in 101	1 in 204
6	OSTM1	Osteopetrosis, autosomal recessive type 5	< 1 in 500	Reduced
16	OTOA	Deafness, autosomal recessive, type 22	1 in 500	1 in 1667
2	OTOF	Deafness, autosomal recessive, type 9	1 in 228	1 in 22,701
1	P3H1	Osteogenesis imperfecta, type 8	1 in 567	1 in 1,447
12	PAH	Phenylketonuria	1 in 60	1 in 857
20	PANK2	Neurodegeneration with brain iron accumulation type 1	1 in 400	1 in 5000
11	PC	Pyruvate carboxylase deficiency	1 in 251	1 in 3150
13	PCCA	Propionic acidemia	1 in 636	1 in 2544
3	PCCB	Propionic acidemia	1 in 635	1 in 7938
10	PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	1 in 497	1 in 1657
5	PDE6A	Retinitis pigmentosa type 43	1 in 500	1 in 863
4	PDE6B	Retinitis pigmentosa type 40	1 in 200	1 in 4000
10	PDE6C	Cone dystrophy type 4	N/A	N/A
17	PDE6G	Retinitis pigmentosa type 57	< 1 in 500	Reduced
8	PDP1	Pyruvate dehydrogenase phosphatase deficiency	< 1 in 500	Reduced
10	PDSS1	Coenzyme Q10 deficiency, primary, type 2	< 1 in 500	Reduced
6	PDSS2	Coenzyme Q10 deficiency, primary, type 3	< 1 in 500	Reduced
13	PDX1	Pancreatic agenesis type 1	< 1 in 500	Reduced
10	PDZD7	Deafness, autosomal recessive, type 57; Usher syndrome, type 2C, digenic	N/A	N/A
7	PEX1	Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)	1 in 191	1 in 3820
17	PEX12	Peroxisome biogenesis disorder type 3A (Zellweger)	< 1 in 500	Reduced
8	PEX2	Peroxisome biogenesis disorder type 5A (Zellweger)	< 1 in 500	Reduced
22	PEX26	Peroxisome biogenesis disorder type 7A (Zellweger)	< 1 in 500	Reduced
12	PEX5	Peroxisome biogenesis disorder type 2A (Zellweger)	< 1 in 500	Reduced
6	PEX7	Rhizomelic chondrodysplasia punctata, type 1	1 in 371	1 in 7420
1	PGM1	Congenital disorder of glycosylation, type 1t	N/A	N/A
16	PHKG2	Glycogen storage disease type 9c	N/A	N/A
10	PHYH	Refsum disease	< 1 in 500	Reduced
2	PJVK	Deafness, autosomal recessive, type 59	N/A	N/A
6	PKHD1	Polycystic kidney disease type 4	1 in 66	1 in 264

1	PKLR	Pyruvate kinase deficiency	1 in 160	1 in 3200
22	PLA2G6	Infantile neuroaxonal dystrophy type 1	1 in 343	1 in 856
10	PLCE1	Nephrotic syndrome, type 3	< 1 in 500	Reduced
8	PLEC	Epidermolysis bullosa simplex with muscular dystrophy	N/A	N/A
1	PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate, type C	N/A	N/A
6	PLG	Plasminogen deficiency, type I	< 1 in 500	Reduced
1	PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	1 in 159	1 in 299
16	PMM2	Congenital disorder of glycosylation, type 1A	1 in 71	1 in 3550
17	PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	1 in 1,107	1 in 3,983
15	POLG	POLG-related disorders	1 in 194	1 in 340
1	POMGNT1	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])	1 in 315	1 in 31500
9	POMT1	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy type 11 [LGMD R11])	1 in 372	1 in 1,708
14	POMT2	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	< 1 in 500	Reduced
3	POU1F1	Pituitary hormone deficiency, combined, type 1	1 in 32	1 in 126
1	PPT1	Ceroid lipofuscinosis, neuronal, type 1	1 in 488	1 in 4880
17	PRCD	Retinitis pigmentosa, type 36	N/A	N/A
2	PRKRA	Dystonia, type 16	< 1 in 500	Reduced
4	PROM1	Retinitis pigmentosa, type 41	1 in 323	1 in 6460
5	PROP1	Pituitary hormone deficiency, combined, type 2	1 in 84	1 in 4200
19	PRX	Charcot-Marie-Tooth disease, type 4F	N/A	N/A
10	PSAP	Combined SAP deficiency	< 1 in 500	Reduced
9	PSAT1	Neu-Laxova syndrome, type 2	N/A	N/A
11	PYGM	McArdle disease	1 in 206	1 in 2060
6	RAB23	Carpenter syndrome	< 1 in 500	Reduced
15	RAB27A	Griscelli syndrome, type 2	N/A	N/A
2	RAB3GAP1	Warburg micro syndrome, type 1	N/A	N/A
1	RAB3GAP2	Martsolf syndrome 1; Warburg micro syndrome 2	N/A	N/A
11	RAG1	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	1 in 344	1 in 614
11	RAG2	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	1 in 1,925	1 in 19250
11	RAPSN	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	1 in 165	1 in 1650
18	RAX	Isolated microphthalmia, type 3	1 in 159	1 in 475
14	RDH12	Leber congenital amaurosis, type 13	1 in 456	1 in 4560
11	RDX	Deafness, autosomal recessive, type 24	< 1 in 500	Reduced
7	RELN	Lissencephaly 2 (Norman-Roberts type)	N/A	N/A
1	REN	Renal tubular dysgenesis	< 1 in 500	Reduced
3	RHO	Retinitis pigmentosa, type 4; Retinitis punctata albescens	1 in 416	1 in 8320
15	RLBP1	Bothnia retinal dystrophy; Fundus albipunctatus	< 1 in 500	Reduced
1	RPE65	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy	1 in 366	1 in 4575
16	RPGRIP1L	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	1 in 319	1 in 860
19	RYR1	Minicore myopathy with external ophthalmoplegia	< 1 in 500	Reduced
13	SACS	Spastic ataxia, Charlevoix-Saguenay, type	1 in 100	1 in 1000
2	SAG	Oguchi disease, type 1	< 1 in 500	Reduced
7	SBDS	Shwachman-Diamond syndrome	1 in 224	1 in 804
11	SBF2	Charcot-Marie-Tooth disease, type 4B2	N/A	N/A
11	SC5D	Lathosterolosis	< 1 in 500	Reduced
12	SCNN1A	Pseudohypoaldosteronism, type 1	N/A	N/A
16	SCNN1B	Pseudohypoaldosteronism, type 1	< 1 in 500	Reduced
16	SCNN1G	Pseudohypoaldosteronism, type 1	< 1 in 500	Reduced
1	SELENON	Muscular dystrophy, rigid spine, type 1	N/A	N/A
1	SEMA4A	Cone-rod dystrophy, type 10; Retinitis pigmentosa, type 35	N/A	N/A
9	SETX	Spinocerebellar ataxia, autosomal recessive, type 1	1 in 500	1 in 2273
17	SGCA	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	1 in 288	1 in 1920
4	SGCB	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	1 in 628	1 in 2093
13	SGCG	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	1 in 1,132	1 in 5,468
17	SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo A)	1 in 253	1 in 5060
5	SH3TC2	Charcot-Marie-Tooth disease, type 4C	1 in 130	1 in 1300
5	SIL1	Marinesco-Sjogren syndrome	< 1 in 500	Reduced
14	SIX6	Optic disc anomalies with retinal and/or macular dystrophy	< 1 in 500	Reduced
15	SLC12A1	Bartter syndrome, type 1	< 1 in 500	Reduced
15	SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	< 1 in 500	Reduced
6	SLC17A5	Salla disease	1 in 328	1 in 2187
15	SLC24A1	Night blindness, congenital stationary (complete), type 1D, autosomal recessive	< 1 in 500	Reduced
7	SLC25A13	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	1 in 619	1 in 2063
13	SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	< 1 in 500	Reduced
11	SLC25A22	Epileptic encephalopathy, early infantile, type 3	< 1 in 500	Reduced
5	SLC26A2	Achondrogenesis Ib; Atelosteogenesis, type II;De la Chapelle dysplasia; Diastrophic dysplasia;Diastrophic dysplasia, broad bone-platyspondylic variant;Epiphyseal dysplasia, multiple, 4	1 in 129	1 in 4300

7	SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome	1 in 88	1 in 587
7	SLC26A5	?Deafness, autosomal recessive, type 61	N/A	N/A
6	SLC35A1	Congenital disorder of glycosylation, type 2F	< 1 in 500	<1 in 1,500
11	SLC35C1	Congenital disorder of glycosylation, type 2C	< 1 in 500	Reduced
1	SLC35D1	Schneckenbecken dysplasia	< 1 in 500	Reduced
11	SLC37A4	Glycogen storage disease, type 1B	1 in 500	1 in 7143
5	SLC45A2	Albinism, oculocutaneous, type 4	< 1 in 500	<1 in 1,600
20	SLC4A11	Corneal endothelial dystrophy, autosomal recessive	1 in 295	1 in 1475
5	SMN1	Spinal muscular atrophy	1 in 36	1 in 360
11	SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B	1 in 350	1 in 3500
22	SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	< 1 in 500	Reduced
13	SPART	Spactic paraplegia, type 20, autosomal recessive	< 1 in 500	Reduced
15	SPG11	Amyotrophic lateral sclerosis 5, juvenile; Charcot-Marie-Tooth disease, axonal, type 2X; Spastic paraplegia 11	1 in 192	1 in 467
16	SPG7	Spastic paraplegia, type 7, autosomal recessive	1 in 80	1 in 183
8	STAR	Lipoid adrenal hyperplasia	1 in 1,147	1 in 14338
1	STIL	Microcephaly, type 7, primary, autosomal recessive	N/A	N/A
15	STRA6	Microphthalmia, isolated, with coloboma, type 8	N/A	N/A
15	STRC	Deafness, autosomal recessive, type 16	1 in 68	1 in 80
2	SUCLG1	Mitochondrial DNA depletion syndrome, type 9 (encephalomyopathic, type with methylmalonic aciduria)	N/A	N/A
12	SUOX	Sulfite oxidase deficiency	< 1 in 500	Reduced
16	TAT	Tyrosinemia, type 2	< 1 in 500	Reduced
1	TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy; Hypoparathyroidism-retardation-dysmorphism syndrome Kenny-Caffey syndrome type 1	N/A	N/A
17	TCAP	Limb-girdle muscular dystrophy, type 7 (LGMD R7)	< 1 in 500	Reduced
11	TCIRG1	Osteopetrosis, autosomal recessive, type 1	1 in 399	1 in 7980
11	TECTA	Deafness, autosomal recessive, type 21	N/A	N/A
5	TERT	Dyskeratosis congenita, autosomal recessive, type 4	< 1 in 500	Reduced
7	TFR2	Hemochromatosis, type 3	< 1 in 500	Reduced
11	TH	Segawa syndrome, recessive	< 1 in 500	Reduced
16	TK2	Mitochondrial DNA depletion syndrome , type 2 (myopathic type)	1 in 500	1 in 16667
9	TMC1	Deafness, autosomal recessive, type 7	N/A	N/A
11	TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2	< 1 in 500	Reduced
8	TMEM67	Meckel syndrome 3; COACH syndrome 1; Joubert syndrome 6; Nephronophthisis 11	1 in 147	1 in 2,940
3	TMIE	Deafness, autosomal recessive, type 6	< 1 in 500	Reduced
21	TMPRSS3	Deafness, autosomal recessive, type 8/10	1 in 135	1 in 2700
19	TNNT1	Nemaline myopathy , type 5, Amish type	< 1 in 500	Reduced
11	TPP1	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	1 in 266	1 in 1773
9	TPRN	Deafness, autosomal recessive, type 79	N/A	N/A
3	TREX1	Aicardi-Goutieres syndrome, type 1	1 in 98	1 in 186
9	TRIM32	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	1 in 226	1 in 376
17	TRIM37	Mulibrey nanism	< 1 in 500	Reduced
22	TRIOBP	Deafness, autosomal recessive, type 28	1 in 445	1 in 8900
17	TSEN54	Pontocerebellar hypoplasia, type 2A; Pontocerebe lar hypoplasia, type 4	1 in 223	1 in 3,997
12	TSFM	Combined oxidative phosphorylation deficiency, type 3	< 1 in 500	Reduced
1	TSHB	Hypothyroidism, congenital, nongoitrous, type 4	1 in 62	1 in 306
14	TSHR	Hypothyroidism, congenital, nongoitrous, type 1	1 in 62	1 in 189
2	TTN	Limb-girdle muscular dystrophy type 10 (LGMDR10); Early-onset myopathy with fatal cardiomyopathy (Salih myopathy)	< 1 in 500	Reduced
8	TTPA	Ataxia with isolated vitamin E deficiency	< 1 in 500	Reduced
6	TULP1	Retinitis pigmentosa 14; Leber congenital amaurosis 15	N/A	N/A
10	TWNK	Mitochondrial DNA depletion syndrome, type 7 (hepatocerebral type); Perrault syndrome type 5	< 1 in 500	Reduced
11	TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	1 in 92	1 in 1840
9	TYRP1	Albinism, oculocutaneous, type 3	< 1 in 500	<1 in 1,400
15	UBR1	Johanson-Blizzard syndrome	N/A	N/A
2	UGT1A1	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	1 in 500	1 in 5,496
5	UQCRQ	Mitochondrial complex III deficiency, nuclear, type 4	< 1 in 500	Reduced
11	USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	1 in 257	1 in 3671
17	USH1G	Usher syndrome, type 1G	< 1 in 500	Reduced
1	USH2A	Usher syndrome, type 2A; Retinitis pigmentosa 39	1 in 70	1 in 467
12	VDR	Rickets, vitamin D-resistant, type 2A	N/A	N/A
9	VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion, type 1	< 1 in 500	Reduced
9	VPS13A	Choreoacanthocytosis	1 in 341	1 in 974
15	VPS33B	Arthrogryposis, renal dysfunction and cholestasis, type 1	N/A	N/A
19	WDR62	Microcephaly, type 2, primary, autosomal recessive, with or without cortical malformations	N/A	N/A
4	WFS1	Wolfram syndrome, type 1	1 in 370	1 in 3700
9	WHRN	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	1 in 93	1 in 127
2	WNT10A	WNT10A-related conditions	1 in 238	1 in 2975
3	WNT7A	Fuhrmann syndrome	< 1 in 500	Reduced
9	XPA	Xeroderma pigmentosum, group A	< 1 in 500	Reduced

14	ZFYVE26	Spastic paraplegia, type 15, autosomal recessive	< 1 in 500	Reduced
1	ZMPSTE24	Mandibuloacral dysplasia with, type B lipodystrophy	N/A	N/A
16	ZNF469	Brittle cornea syndrome, type 1	N/A	N/A

N/A: no data prevalence unknown