CGT Exome v3.3.4

Patient Information		Sample Information		Clinic Information	
Unique pat id.:	0059120 - 15431234	**Sample type:**	Blood (EDTA)	**Clinic:**	IVI BUENOS AIRES
Patient name:		**Date of draw:**	24/11/2022	**Clinician:**	FERNANDO NEUSPILLER
Patient DOB:		**Date of receipt:**	01/12/2022		
Gender:	Female	**Report date/time:**	02/02/2023 12:08		
Ethnic group:	Caucasian				
Indication:	No family history				

TEST RESULTS

POSITIVE

The individual is carrier of:

Bardet-Biedl syndrome, type 12

Gene :	BBS12	**Allele:**	Het
DNA Change:	NM_001178007.1:c.2023C>T	**Inheritance:**	AR
Protein change:	p.Arg675*	**OMIM phenotype:**	615989
ACMG classification:	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 is also the case for a negative personal result when a partner or a gamete donor is a carrier for one or more of these analyzed genes. However, due to test limitations associated with any genetic test, this low risk is not zero (see limitations section and informed consent)

LOW COVERAGE VARIANTS

NAGLU:NM_000263.3:c.245G>A;RYR1:NM_000540.2:c.13068_13078delCCTGCTCTGGG;PLAA:NM_001031689.2:c.1049A>T;ZNF469:NM_001127464.2:c.3034delG;NEK1:NM_001199397.1:c.3616G>T;TTN:NM_001267550.2:c.11254+2T>C;NTRK1:NM_002529.3:c.1A>C;GCSH:NM_004483.4:c.1A>G;LZTR1:NM_006767.3:c.1005_1012delTGAAGTGC;LZTR1:NM_006767.3:c.1018C>T;LZTR1:NM_006767.3:c.1030delT;LZTR1:NM_006767.3:c.1084C>T;LZTR1:NM_006767.3:c.1107delT;FOXE3:NM_012186.2:c.720C>A;CRIPT:NM_014171.5:c.132delA;CRIPT:NM_014171.5:c.133_134insGG;PIEZO2:NM_022068.3:c.4169_4170delCA;FA2H:NM_024306.4:c.70dupG;PKHD1:NM_138694.3:c.9856_9859dupAGTT.These variants have a coverage lower than 7X and it is not possible to determine if they are present or not in the sample (non-informative variants).

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) and 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

English language.

TEST METHODOLOGY

1.DNA extraction from the biological sample. 2. Next Generation Sequencing of gene regions where known pathogenic variants are located (list available at https://cgt.igenomix.com/diseases-list/). 3. Raw data analysis using bioinformatics (bioinformatic pipeline v1.1). QC parameters require that more than 99.7% of the tested variants have coverage greater than the minimum read depth (7x). 4. Complementary testing by other techniques for: a) the SMN1 gene: exon 7 deletion; b) the CYP21A2 gene: frequent mutations; c) HBA1/HBA2 genes: frequent deletions; d) the FMR1 gene: CGG repeat sizing (females only); e) the DMD gene: frequent deletions/duplications; f) the F8 gene: intron 22 inversion (females only); g) the FXN gene: GAA repeat.

TEST LIMITATIONS

The CGT test only includes analysis of the specific variants included in the list (list of variants analysed are available by request), and no others. Therefore, the CGT test does not cover all monogenic diseases nor 100% of disease-causing variants for each tested gene. The test does not include the analysis of conditions associated with mitochondrial DNA, multifactorial, digenic or dominant inheritance. The test does not detect large rearrangements (inversions, deletions and duplications more than 15 nucleotides), pathogenic variants located in regulatory regions or intronic regions outside the +/-3bp cut off or in low sequence coverage areas. 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 be certain that the two copies are each in one of the two alleles (non-carrier) or if both are in the same allele (cis) and no copies in the other (silent carrier). Finally, if our assessment of a variant fails to meet our QC parameters due to low coverage, a result for the variant(s) will not be issued.
The analytical detection rate is higher than 99%. The clinical sensitivity varies among conditions (e.g.: for the HEXB gene, 30% of affected patients are carriers of a 16 kb deletion that is not included in the test). The sensitivity for SMN1 is approximately 96% because point mutations or small insertions/deletions are not analyzed and, for a normal result (2 copies detected), it is not possible to be certain that the two copies are each in one of the two alleles (non-carrier) or if both are in the same allele (cis) and no copies in the other (carrier).
A negative result for the variants included in the CGT test does not exclude the possibility of being a carrier. The presence of pseudogenes and/or rare polymorphisms and/or homopolymers may lead to false negative or false positive results. A negative result for the CGT variants does not exclude the possibility of a de novo pathogenic variant being present in the offspring. 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. 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 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

IGENOMIX ARGENTINA S.A will only release the report once a completed Test Requisition Form is received. The clinic/clinician/certified health professional requesting the test is responsible for obtaining and taking custody of "Informed Consent" from the patient as depicted by national guidelines and/or legislation.
This test was developed, and its performance characteristics determined by IGENOMIX ARGENTINA S.A . 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.
*IGENOMIX SPAIN holds CLIA Certificate of Compliance: #99D2146167. Part of this test has been outsourced to a referral laboratory whose QMS is based on high Quality Standards, periodically monitored by Igenomix SPAIN and audited by independent external parties.

EXEMPTION CLAUSE OF DIAGNOSTIC LIABILITY

The genetic diagnosis services carried out by IGENOMIX ARGENTINA S.A 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 ARGENTINA S.A is not responsible for any decisions made or actions undertaken by the contracting party based on the results provided by IGENOMIX ARGENTINA S.A 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

Ana Cervero PhD
2703-CV
Laboratory Director

COUNTERSIGNED

Lic. Daniela Lorenzi

Manager de Laboratorio

This test or part of this test has been outsourced to a referral Laboratory (IGENOMIX Group) CLIA #99D2146167

Bardet-Biedl syndrome, type 12

What is Bardet-Biedl syndrome, type 12?

Bardet-Biedl syndrome 12 (BBS12) is a clinically pleiotropic autosomal recessive ciliopathy. The patients with BBS12 studied by Stoetzel et al. (2007) and Harville et al. (2010) met the diagnostic criteria of Beales et al. (1999), which required the presence of either 4 primary features, including rod-cone dystrophy, polydactyly, obesity, learning disabilities, hypogonadism (in males), and/or renal anomalies; or 3 primary plus 2 secondary features (e.g., developmental delay, ataxia, cataracts).

What is the next step if I am a carrier of Bardet-Biedl syndrome, type 12?

If you are a carrier of Bardet-Biedl syndrome, type 12 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?

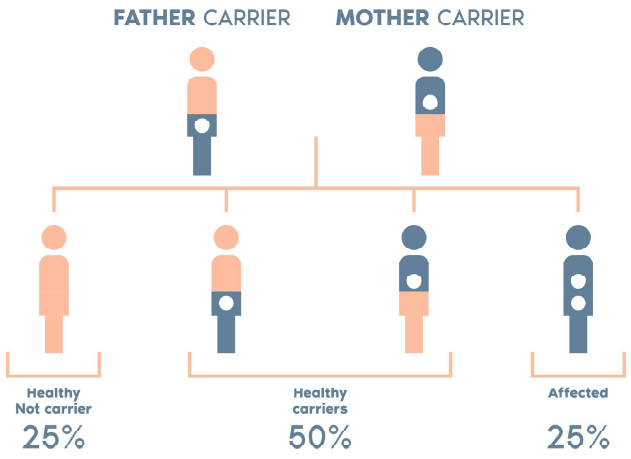
If your partner tests negative for Bardet-Biedl syndrome, type 12, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Bardet-Biedl syndrome, type 12?

When both parents are carriers of Bardet-Biedl syndrome, type 12, 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

AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA12, ABCA3, ABCA4, ABCB11, ABCB4, ABCC2, ABCC6, ABCC8, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACO2, ACOX1, ACOX2, ACP5, ACSF3, ACTA1, ACY1, ADA, ADA2, ADAM9, ADAMTS10, ADAMTS13, ADAMTS17, ADAMTS18, ADAMTS2, ADAMTSL2, ADAMTSL4, ADAR, ADAT3, ADGRG1, ADGRG6, ADGRV1, ADK, ADSL, ADSS1, AFG3L2, AFP, AGA, AGBL5, AGK, AGL, AGPAT2, AGPS, AGRN, AGT, AGTR1, AGXT, AHCY, AHI1, AICDA, AIMP1, AIMP2, AIPL1, AIRE, AK1, AK2, AKR1C2, AKR1D1, ALAD, ALB, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX12B, ALOXE3, ALPK3, ALPL, ALS2, ALX1, ALX3, ALX4, AMACR, AMBN, AMH, AMHR2, AMN, AMPD1, AMPD2, AMT, ANGPTL3, ANKS6, ANO10, ANO5, ANTXR1, ANTXR2, AP1S1, AP1S2, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOC2, APOE, APRT, APTX, AQP2, AR, ARFGEF2, ARG1, ARHGDIA, ARHGEF18, ARL13B, ARL2BP, ARL6, ARMC9, ARPC1B, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASL, ASNS, ASPA, ASPH, ASPM, ASS1, ATAD1, ATF6, ATIC, ATM, ATOH7, ATP13A2, ATP2A1, ATP6V0A2, ATP6V0A4, ATP6V1A, ATP6V1B1, ATP6V1E1, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, AURKC, AVIL, B2M, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAT2, BCHE, BCKDHA, BCKDHB, BCKDK, BCL10, BCS1L, BEST1, BFSP1, BHLHA9, BIN1, BLM, BLNK, BLOC1S3, BLOC1S6, BLVRA, BMP1, BMPER, BMPR1B, BOLA3, BPGM, BPNT2, BRAT1, BRF1, BRIP1, BRWD3, BSCL2, BSND, BTD, BTK, BUB1B, C12orf57, C12orf65, C19orf12, C1QA, C1QB, C1QBP, C1QC, C1S, C2, C2CD3, C3, C5, C6, C7, C8B, C8orf37, CA12, CA2, CA5A, CA8, CABP2, CABP4, CACNA1D, CACNA2D4, CAD, CALCRL, CANT1, CAPN1, CAPN3, CARD11, CARD9, CARS2, CASQ2, CASR, CAST, CAT, CATSPER1, CAVIN1, CBLIF, CBS, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC115, CCDC174, CCDC39, CCDC40, CCDC65, CCDC8, CCDC88C, CCN6, CCNO, CD19, CD247, CD27, CD2AP, CD320, CD36, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD59, CD79A, CD79B, CD81, CD8A, CDAN1, CDC14A, CDC45, CDCA7, CDH11, CDH23, CDH3, CDHR1, CDIN1, CDK10, CDK5RAP2, CDSN, CDT1, CEBPE, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP19, CEP290, CEP41, CEP55, CEP57, CEP78, CEP83, CERKL, CERS3, CFAP43, CFAP53, CFD, CFH, CFI, CFL2, CFTR, CHAT, CHKB, CHM, CHMP1A, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHST6, CHSY1, CHUK, CIB2, CIITA, CILK1, CISD2, CIT, CKAP2L, CLCF1, CLCN1, CLCN2, CLCN7, CLCNKA, CLCNKB, CLDN1, CLDN10, CLDN14, CLDN16, CLDN19, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM2, CNNM4, CNPY3, CNTNAP1, CNTNAP2, COA6, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A1, COL11A2, COL13A1, COL17A1, COL18A1, COL1A2, COL25A1, COL27A1, COL4A3, COL4A4, COL4A5, COL6A1, COL6A2, COL6A3, COL7A1, COL9A1, COL9A2, COLEC10, COLEC11, COLQ, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CP, CPA6, CPAMD8, CPLANE1, CPLX1, CPS1, CPT1A, CPT2, CR2, CRADD, CRB1, CRB2, CRBN, CRIPT, CRLF1, CRPPA, CRTAP, CRYAA, CRYAB, CRYBB1, CRYBB3, CSF2RB, CSF3R, CSPP1, CSTA, CSTB, CTC1, CTH, CTNS, CTPS1, CTSA, CTSC, CTSD, CTSF, CTSK, CUBN, CUL4B, CUL7, CWC27, CWF19L1, CYB5A, CYB5R3, CYBA, CYBB, CYC1, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP21A2, CYP24A1, CYP26B1, CYP26C1, CYP27A1, CYP27B1, CYP2R1, CYP2U1, CYP4F22, CYP4V2, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBH, DBT, DCAF17, DCC, DCDC2, DCHS1, DCLRE1C, DCPS, DCX, DDB2, DDC, DDHD1, DDHD2, DDR2, DDRGK1, DDX11, DDX59, DENND5A, DES, DGAT1, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHH, DHODH, DHPS, DHTKD1, DIAPH1, DIS3L2, DKC1, DLAT, DLD, DLG3, DLL3, DMD, DMGDH, DMP1, DMXL2, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAJB2, DNAJC12, DNAJC19, DNAJC21, DNAJC6, DNAL1, DNASE1L3, DNM1L, DNM2, DNMT3B, DOCK2, DOCK6, DOCK7, DOCK8, DOK7, DOLK, DONSON, DPAGT1, DPH1, DPM1, DPM2, DPM3, DPY19L2, DPYD, DPYS, DRAM2, DRC1, DSG1, DSG4, DSP, DST, DSTYK, DTNBP1, DUOX2, DUOXA2, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, DYSF, DZIP1L, EARS2, ECEL1, ECHS1, ECM1, EDA, EDAR, EDARADD, EDN1, EDN3, EDNRB, EFEMP2, EFL1, EGFR, EGR2, EIF2AK3, EIF2AK4, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4A3, ELAC2, ELMO2, ELOVL4, ELP1, ELP2, EMC1, EMD, EML1, EMP2, ENAM, ENO3, ENPP1, ENTPD1, EOGT, EPB41, EPB42, EPCAM, EPG5, EPM2A, EPRS1, EPS8L2, ERAL1, ERBB3, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERLIN1, ERLIN2, ESCO2, ESPN, ESR1, ESRRB, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EXPH5, EXT1, EXTL3, EYS, F10, F11, F13A1, F13B, F2, F5, F7, F8, F9, FA2H, FADD, FAH, FAM126A, FAM161A, FAM20A, FAM20C, FAN1, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAR1, FARS2, FASTKD2, FAT4, FBLN5, FBP1, FBXL4, FBXO7, FDXR, FECH, FERMT1, FERMT3, FEZF1, FGA, FGB, FGD1, FGD4, FGF23, FGF3, FGG, FH, FIBP, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLG, FLI1, FLNB, FLVCR1, FLVCR2, FMN2, FMO3, FMR1, FOLR1, FOXE1, FOXE3, FOXN1, FOXRED1, FRAS1, FREM1, FREM2, FRRS1L, FSHB, FSHR, FTCD, FTL, FTO, FTSJ1, FUCA1, FUT8, FXN, FYCO1, FZD6, G6PC, G6PC3, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALNT3, GALT, GAMT, GAN, GAS8, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCK, GCM2, GCNT2, GCSH, GDAP1, GDF1, GDF5, GDF6, GFER, GFM1, GFPT1, GGCX, GH1, GHR, GHRHR, GHSR, GINS1, GIPC3, GJA1, GJB1, GJB2, GJB6, GJC2, GLA, GLB1, GLDC, GLDN, GLE1, GLIS2, GLIS3, GLRA1, GLRB, GLRX5, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAT1, GNAT2, GNB5, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNRHR, GNS, GORAB, GOSR2, GOT2, GP1BA, GP1BB, GP6, GP9, GPAA1, GPC6, GPD1, GPHN, GPI, GPIHBP1, GPR143, GPR179, GPR68, GPSM2, GPT2, GPX4, GRHL2, GRHPR, GRID2, GRIK2, GRIN1, GRIP1, GRK1, GRM1, GRM6, GRN, GRXCR1, GSC, GSS, GTF2H5, GTPBP2, GTPBP3, GUCY2C, GUCY2D, GUF1, GUSB, GYG1, GYS1, GYS2, GZF1,

H6PD, HAAO, HACE1, HADH, HADHA, HADHB, HAMP, HARS1, HAX1, HBA, HBA1, HBA2, HBB, HCFC1, HELLS, HEPACAM, HERC1, HERC2, HES7, HESX1, HEXA, HEXB, HFM1, HGD, HGF, HGSNAT, HIBCH, HIKESHI, HINT1, HJV, HK1, HLCS, HMGCL, HMGCS2, HMOX1, HMX1, HNMT, HOGA1, HOXA1, HOXB1, HOXC13, HPCA, HPD, HPGD, HPRT1, HPS1, HPS3, HPS4, HPS5, HPS6, HPSE2, HR, HSD11B2, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, HTRA1, HTRA2, HYAL1, HYDIN, HYLS1, IARS1, IBA57, ICOS, IDH3B, IDS, IDUA, IER3IP1, IFNGR1, IFNGR2, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IGF1, IGF1R, IGFALS, IGFBP7, IGHMBP2, IGLL1, IHH, IKBKB, IL10RA, IL10RB, IL11RA, IL12B, IL12RB1, IL17RA, IL17RC, IL1RAPL1, IL1RN, IL21R, IL2RA, IL2RG, IL36RN, IL7R, ILDR1, IMPA1, IMPG2, INPP5E, INPP5K, INPPL1, INS, INSR, INTS1, INVS, IQCB1, IQCE, IRAK4, IRF8, IRX5, ISCA1, ISCA2, ISCU, ISG15, ITCH, ITGA2B, ITGA3, ITGA6, ITGA7, ITGA8, ITGB2, ITGB3, ITGB4, ITGB6, ITK, ITPA, ITPR1, IVD, IYD, JAGN1, JAK3, JAM3, JUP, KANK2, KARS1, KATNB1, 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You can review the list of pathogenic variants analysed for the above genes on our website: https://cgt.igenomix.com/diseases-list. If you are unable to access this list digitally, please contact our customer support department for more information.

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).

Gene	Disease/Condition	Carrier Rate	Residual Risk
AAAS	Triple-A syndrome (achalasia-addisonianism-alacrimia)	1 in 436	1 in 8,266
ABCA12	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	1 in 194	1 in 715
ABCA3	Surfactant metabolism dysfunction, pulmonary, type 3	1 in 500	1 in 7,143
ABCA4	Stargardt disease type 1; Cone-rod dystrophy type 3	1 in 62	1 in 119
ABCB11	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	1 in 276	1 in 614
ABCC8	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	1 in 192	1 in 1,690
ABCD1	Adrenoleukodystrophy	1 in 14,000	1 in 28,579
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	1 in 496	1 in 49,501
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	N/A	N/A
ACAD9	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	1 in 309	1 in 576
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	1 in 50	<1 in 488
ACADS	Short-chain acyl-CoA dehydrogenase deficiency	1 in 102	1 in 1,015
ACADSB	Short/branched-chain acyl-CoA dehydrogenase deficiency	1 in 500	1 in 1,125
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	1 in 112	1 in 698
ACAT1	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	1 in 500	1 in 769
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	1 in 2,394	1 in 7,180
ACSF3	Combined malonic and methylmalonic aciduria	1 in 67	1 in 90
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	1 in 390	1 in 2,335
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	1 in 2,432	1 in 4,053
ADK	Hypermethioninemia due to adenosine kinase deficiency	1 in 500	1 in 1,498
AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)	1 in 975	1 in 3,442
AGL	Glycogen storage disease, type 3	1 in 259	1 in 1,083
AGPS	Rhizomelic chondrodysplasia punctata, type 3	1 in 18,591	<1 in 1,000,000
AGXT	Hyperoxaluria, primary, type 1	1 in 174	>1 in 5,758
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	1 in 708	1 in 2,122
AHI1	Joubert syndrome, type 3	1 in 334	1 in 706
AIPL1	Leber congenital amaurosis, type 4	1 in 160	1 in 189
AIRE	Autoimmune polyendocrinopathy syndrome, type 1	1 in 209	1 in 1,665
ALDH3A2	Sjogren-Larsson syndrome	1 in 718	1 in 4,231
ALDH4A1	Hyperprolinemia, type 2	1 in 500	1 in 49,951
ALDOB	Fructose intolerance, hereditary	1 in 67	1 in 298
ALG1	Congenital disorder of glycosylation, type 1K	1 in 87	1 in 130
ALG6	Congenital disorder of glycosylation, type 1C	1 in 301	1 in 421
ALMS1	Alström syndrome	1 in 168	1 in 488
ALPL	Hypophosphatasia, infantile/childhood	1 in 274	1 in 1,348
AMT	Glycine encephalopathy	1 in 779	1 in 3,891
ANO10	Spinocerebellar ataxia, autosomal recessive, type 10	1 in 224	1 in 2,236
AP1S2	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	1 in 500,000	1 in 1,000,000
AQP2	Diabetes insipidus, nephrogenic, type 2	1 in 721	1 in 1,773
AR	Androgen insensitivity syndrome, complete	1 in 16,639	1 in 37,670

ARG1	Argininemia (arginase deficiency)	1 in 2,559	1 in 6,745
ARL13B	Joubert syndrome type 8	1 in 72	1 in 119
ARSA	Metachromatic leukodystrophy	1 in 135	1 in 2,686
ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	1 in 314	1 in 1,023
ARX	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	1 in 37,038	1 in 64,815
ASL	Argininosuccinic aciduria	1 in 117	1 in 372
ASNS	Asparagine synthetase deficiency	1 in 2,023	1 in 2,567
ASPA	Canavan disease	1 in 486	1 in 1,458
ASS1	Citrullinemia, type 1	1 in 323	1 in 1,124
ATM	Ataxia-telangiectasia	1 in 150	1 in 675
ATP6V1B1	Renal tubular acidosis with deafness	1 in 1,092	1 in 2,401
ATP7A	Menkes disease; Occipital horn syndrome	1 in 150,000	1 in 501,722
ATP7B	Wilson disease	1 in 42	1 in 249
ATP8B1	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1	1 in 738	1 in 1,803
ATRX	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/mental retardation syndrome	1 in 500,000	1 in 919,360
AUH	3-methylglutaconic aciduria, type 1	<1 in 500	<1 in 938
B4GALT1	Congenital disorder of glycosylation, type 2D	<1 in 500	<1 in 50,000
BBS1	Bardet-Biedl syndrome, type 1	1 in 152	1 in 490
BBS10	Bardet-Biedl syndrome, type 10	1 in 237	1 in 666
BBS12	Bardet-Biedl syndrome, type 12	1 in 613	1 in 798
BBS2	Bardet-Biedl syndrome, type 2	1 in 333	1 in 3,255
BCHE	Butyrylcholinesterase deficiency	1 in 30	<1 in 270
BCKDHA	Maple syrup urine disease, type 1A	1 in 555	1 in 2,317
BCKDHB	Maple syrup urine disease, type 1B	1 in 306	1 in 990
BCS1L	Mitochondrial complex III deficiency nuclear type 1; GRACILE syndrome; Bjornstad syndrome	1 in 385	1 in 1,230
BLM	Bloom syndrome	1 in 358	1 in 35,701
BRWD3	Mental retardation, X-linked, type 93	1 in 45,000	1 in 65,000
BSND	Bartter syndrome, type 4A	1 in 916	1 in 2,014
BTD	Biotinidase deficiency	1 in 120	1 in 477
BTK	Agammaglobulinemia X-linked, type 1	1 in 126,556	1 in 275,310
CA2	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	<1 in 500	<1 in 1,000
CAPN3	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	1 in 103	1 in 7,498
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, type 2	1 in 51	1 in 98
CBS	Homocystinuria due to cystathionine beta-synthase	1 in 128	1 in 2,541
CC2D2A	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2	1 in 196	1 in 2,800
CCDC88C	Hydrocephalus, congenital, type 1	1 in 500	1 in 7,143
CD40LG	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	1 in 250,000	1 in 532,258
CDH23	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D	1 in 216	1 in 499
CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	1 in 97	1 in 248
CERKL	Retinitis pigmentosa, type 26	1 in 370	1 in 515

CFTR	Cystic fibrosis	1 in 25	1 in 833
CHAT	Myasthenic syndrome, congenital, type 6, presynaptic	1 in 121	1 in 134
CHM	Choroideremia	1 in 33,334	1 in 64,000
CHRNE	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	1 in 244	1 in 491
CHRNG	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	1 in 400	1 in 1,597
CHST6	Macular corneal dystrophy	1 in 80	1 in 394
CIITA	Bare lymphocyte syndrome, type 2, complementation group A	1 in 924	1 in 1,501
CLCN1	Myotonia congenita, recessive	1 in 159	1 in 319
CLN3	Ceroid lipofuscinosis, neuronal, type 3	1 in 242	1 in 2,090
CLN5	Ceroid lipofuscinosis, neuronal, type 5	1 in 762	1 in 3,299
CLN6	Ceroid lipofuscinosis, neuronal, type 6	1 in 977	1 in 2,840
CLN8	Ceroid lipofuscinosis, neuronal, type 8	1 in 1,250	1 in 2,276
CLRN1	Usher syndrome, type 3A	1 in 420	1 in 577
CNGA1	Retinitis pigmentosa type 49	1 in 625	1 in 1,171
CNGB1	Retinitis pigmentosa type 45	1 in 867	1 in 1,614
CNGB3	Achromatopsia, type 3	1 in 87	1 in 1,363
COL27A1	Steel syndrome	1 in 500	N/A
COL4A3	Alport syndrome, autosomal recessive, type 2	1 in 218	1 in 352
COL4A4	Alport syndrome, autosomal recessive, type 2	1 in 349	1 in 565
COL4A5	Alport syndrome, X-linked	1 in 34,622	1 in 72,761
COL7A1	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	1 in 100	1 in 243
COLQ	Myasthenic syndrome, congenital, type 5	1 in 805	1 in 1,420
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 2; Leigh syndrome due to cytochrome c oxidase deficiency	1 in 480	1 in 640
CPS1	Carbamoylphosphate synthetase 1 deficiency	1 in 343	1 in 817
CPT1A	Carnitine palmitoyltransferase type 1A deficiency, hepatic	1 in 1,518	1 in 6,638
CPT2	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	1 in 147	1 in 682
CRB1	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	1 in 158	1 in 203
CRTAP	Osteogenesis imperfecta, type 7	1 in 1,416	1 in 3,539
CTH	Cystathioninuria	1 in 80	1 in 7,870
CTNS	Nephropathic cystinosis	1 in 249	1 in 900
CTSA	Galactosialidosis	1 in 64	1 in 118
CTSC	Haim-Munk syndrome; Papillon-Lefevre syndrome	1 in 500	1 in 2,496
CTSD	Ceroid lipofuscinosis, neuronal, type 10	1 in 1,003	1 in 4,510
CTSK	Pycnodysostosis	1 in 1,067	1 in 3,910
CUL4B	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	1 in 500,000	1 in 636,300
CYBA	Chronic granulomatous disease, type 4	1 in 1,689	1 in 1930
CYBB	Chronic granulomatous disease, X-linked	1 in 100,000	1 in 250,000
CYP11A1	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	1 in 500	1 in 7,143
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	1 in 792	1 in 1,955
CYP11B2	Hypoaldosteronism, congenital, due to CMO I deficiency	1 in 825	1 in 943

CYP17A1	17 alpha(α)-hydroxylase/17,20-lyase deficiency	1 in 560	1 in 679
CYP19A1	Aromatase deficiency	1 in 2,159	1 in 3,532
CYP1B1	Glaucoma, primary congenital, type 3A	1 in 196	1 in 407
CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	1 in 62	1 in 1,228
CYP27A1	Cerebrotendinous xanthomatosis	1 in 275	1 in 1,113
CYP27B1	Vitamin D-dependent rickets, type 1	N/A	N/A
DBT	Maple syrup urine disease, type 2	1 in 410	1 in 40,900
DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	1 in 903	1 in 1,410
DCX	Lissencephaly, X-linked, type 1	1 in 50,000	1 in 158,748
DDB2	Xeroderma pigmentosum, complementation group E	1 in 3,138	1 in 15,686
DHCR7	Smith-Lemli-Opitz syndrome	1 in 70	1 in 1,756
DHDDS	Retinitis pigmentosa, type 59	1 in 6,008	1 in 8,010
DKC1	Dyskeratosis congenita, X-linked	1 in 250,000	1 in 459,999
DLD	Dihydrolipoamide dehydrogenase deficiency	1 in 1,506	1 in 14,549
DLG3	Mental retardation, X-linked, type 90	1 in 45,000	1 in 100,000
DMD	Duchenne/Becker muscular dystrophy	1 in 2,942	1 in 58,819
DNAH5	Ciliary dyskinesia, primary, type 3, with or without situs inversus	1 in 87	1 in 283
DNAI1	Ciliary dyskinesia, primary, type 1, with or without situs inversus	1 in 323	1 in 556
DNAI2	Ciliary dyskinesia, primary, type 9, with or without situs inversus	1 in 758	1 in 1,248
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient	1 in 500	N/A
DOK7	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	1 in 262	1 in 719
DOLK	Congenital disorder of glycosylation, type 1M	<1 in 500	<1 in 563
DPAGT1	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	<1 in 500	<1 in 808
DPM1	Congenital disorder of glycosylation, type 1E	<1 in 500	<1 in 1,750
DPYD	Dihydropyrimidine dehydrogenase deficiency	1 in 558	1 in 55,701
DUOX2	Thyroid dyshormonogenesis, type 6	1 in 80	1 in 170
DUOXA2	Thyroid dyshormonogenesis, type 5	1 in 80	1 in 144
DYNC2H1	Short-rib thoracic dysplasia, type 3, with or without polydactyly	1 in 50	1 in 500
DYSF	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	1 in 199	1 in 1,050
EDA	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	1 in 5,715	1 in 10,610
EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	1 in 131	1 in 252
EIF2AK3	Wolcott-Rallison syndrome	<1 in 500	<1 in 2,500
EIF2B5	Leukoencephalopathy with vanishing white matter (VWM)	1 in 390	1 in 974
EMD	Emery-Dreifuss muscular dystrophy, type 1, X-linked	1 in 88,496	<1 in 1,000,000
ERCC2	Trichothiodystrophy, type 1	1 in 154	1 in 1,378
ERCC3	Trichothiodystrophy, type 2	1 in 436	1 in 1,306
ERCC5	Cerebrooculofacioskeletal syndrome, type 3	1 in 970	1 in 96,901
ERCC6	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	1 in 335	1 in 3,132
ERCC8	Cockayne syndrome, type A	1 in 1,080	1 in 2,670
ESCO2	Roberts syndrome	1 in 1,395	1 in 18,820
ETFA	Glutaric acidemia, type 2A	1 in 857	1 in 2,284
ETFB	Glutaric acidemia, type 2B	1 in 1,593	1 in 2,230

ETFDH	Glutaric acidemia, type 2C	1 in 336	1 in 615
ETHE1	Ethylmalonic encephalopathy	1 in 1,279	1 in 4,794
EVC	Ellis-van Creveld syndrome	1 in 370	1 in 739
EVC2	Ellis-van Creveld syndrome	1 in 240	1 in 718
EXOSC3	Pontocerebellar hypoplasia, type 1B	1 in 139	1 in 300
EYS	Retinitis pigmentosa, type 25	1 in 82	1 in 139
F11	Factor XI deficiency	1 in 500	N/A
F2	Prothrombin deficiency	1 in 415	1 in 1,325
F5	Factor V deficiency	1 in 500	N/A
F8	Hemophilia A	1 in 4,635	<1 in 16,550
F9	Hemophilia B	1 in 15,000	<1 in 29,000
FAH	Tyrosinemia, type 1	1 in 259	1 in 925
FAM161A	Retinitis pigmentosa, type 28	1 in 343	1 in 856
FAM20C	Raine syndrome	<1 in 500	<1 in 1,000
FANCA	Fanconi anemia, complementation group A	1 in 148	1 in 269
FANCC	Fanconi anemia, complementation group C	1 in 431	1 in 1,514
FANCG	Fanconi anemia, complementation group G	1 in 563	1 in 812
FGD1	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	1 in 500000	1 in 937,499
FH	Fumarase deficiency	1 in 252	1 in 1,218
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 310
FKTN	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])	1 in 1,023	1 in 5,841
FMO3	Trimethylaminuria	1 in 100	1 in 1,000
FMR1	Fragile X syndrome	>1 in 368	<1 in 37,000
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19	1 in 439	1 in 822
FRAS1	Fraser syndrome, type 1	1 in 300	1 in 576
FTCD	Glutamate formiminotransferase deficiency	1 in 355	1 in 591
FTSJ1	Mental retardation, X-linked 44	1 in 45,000	N/A
FUCA1	Fucosidosis	1 in 1,149	1 in 4,880
FXN	Friedreich ataxia	1 in 91	1 in 1,014
G6PC	Glycogen storage disease, type 1A	1 in 212	1 in 471
G6PC3	Dursun syndrome	<1 in 500	<1 in 1,170
G6PD	Hemolytic anemia, G6PD deficient (favism)	1 in 25	<1 in 241
GAA	Glycogen storage disease, type 2	1 in 159	1 in 1,577
GALC	Krabbe disease	1 in 67	1 in 120
GALE	Galactose epimerase deficiency	1 in 2,011	1 in 9,381
GALK1	Galactokinase deficiency with cataracts	1 in 747	1 in 1,091
GALNS	Mucopolysaccharidosis, type 4A	1 in 311	1 in 657
GALT	Galactosemia	1 in 102	1 in 2,021
GAMT	Cerebral creatine deficiency syndrome, type 2	1 in 435	1 in 815
GBA	Gaucher disease	1 in 105	1 in 744
GBE1	Glycogen storage disease, type 4	1 in 192	1 in 446

GCDH	Glutaricaciduria, type 1	1 in 100	1 in 1,981
GCH1	Hyperphenylalaninemia, BH4-deficient, type B	1 in 250	1 in 436
GCSH	?Glycine encephalopathy	N/A	N/A
GDAP1	Charcot-Marie-Tooth disease, recessive intermediate, type A	1 in 130	1 in 298
GDF5	Chondrodysplasia, Grebe type	N/A	N/A
GFM1	Combined oxidative phosphorylation deficiency, type 1	1 in 480	1 in 731
GHRHR	Growth hormone deficiency, isolated, type 1B	1 in 50	1 in 83
GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1	1 in 15,161	1 in 44,254
GJB2	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	1 in 40	1 in 489
GJB6	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	1 in 421	1 in 42,000
GLA	Fabry disease	1 in 35,031	<1 in 217,000
GLB1	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	1 in 277	1 in 1,320
GLDC	Glycine encephalopathy	1 in 255	1 in 509
GLE1	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	1 in 453	1 in 582
GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)	1 in 279	1 in 1,593
GNMT	Glycine N-methyltransferase deficiency	1 in 500	1 in 24,951
GNPTAB	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	1 in 176	1 in 17,522
GNPTG	Mucolipidosis III gamma	1 in 684	1 in 2,232
GNRHR	Hypogonadotropic hypogonadism, type 7, without anosmia	1 in 500	1 in 2,097
GNS	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	1 in 2,731	1 in 273,001
GP1BA	Bernard-Soulier syndrome, type A1	1 in 1,677	1 in 3,074
GP1BB	Bernard-Soulier syndrome, type B	1 in 500	1 in 531
GP9	Bernard-Soulier syndrome, type C	1 in 451	1 in 1,576
GPR143	Ocular albinism, type 1 (Nettleship-Falls type)	1 in 42,858	<1 in 48,000
GRHPR	Hyperoxaluria, primary, type 2	1 in 433	1 in 43,201
GRIP1	Fraser syndrome 3	1 in 224	1 in 2,236
GSS	Glutathione synthetase deficiency	1 in 992	1 in 2,974
GUCY2D	Leber congenital amaurosis, type 1	1 in 248	1 in 305
GUSB	Mucopolysaccharidosis, type 7	1 in 552	1 in 1,6531
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency	1 in 415	1 in 622
HADHA	Long-chain 3-hydroxyl-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	1 in 251	1 in 833
HADHB	Mitochondrial trifunctional protein deficiency	1 in 1,379	1 in 3,008
HAX1	Neutropenia, severe congenital, type 3, autosomal recessive	1 in 219	1 in 306
HBA1	Thalassemia, alpha-	1 in 30	1 in 194
HBA2	Thalassemia, alpha-	1 in 30	1 in 194
HBB	Beta-thalassemia, Sickle cell anemia and other HBB-related hemoglobinopathies	1 in 67	1 in 411
HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	1 in 500,000	N/A
HEXA	Tay-Sachs disease	1 in 90	1 in 743
HEXB	Sandhoff disease, infantile, juvenile, and adult forms	1 in 202	1 in 743
HGD	Alkaptonuria	1 in 250	1 in 1,708
HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	1 in 345	1 in 809

HLCS	Holocarboxylase synthetase deficiency	1 in 703	1 in 2,185
HMGCL	HMG-CoA lyase deficiency	1 in 875	1 in 2,623
HMOX1	Heme oxygenase-1 deficiency	1 in 500	N/A
HOGA1	Hyperoxaluria, primary, type 3	1 in 169	1 in 309
HPD	Tyrosinemia, type 3	1 in 2,453	1 in 5,518
HPRT1	Lesch-Nyhan syndrome	1 in 145,204	1 in 382,808
HPS1	Hermansky-Pudlak syndrome, type 1	1 in 493	1 in 1,448
HPS3	Hermansky-Pudlak syndrome, type 3	1 in 491	1 in 589
HSD17B10	HSD10 mitochondrial disease	1 in 500,000	1 in 1,000,000
HSD17B3	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	<1 in 500	<1 in 2,750
HSD17B4	D-bifunctional protein deficiency	1 in 534	1 in 11,727
HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	1 in 862	1 in 1,831
HSPG2	Dyssegmental dysplasia, Silverman-Handmaker type	<1 in 500	<1 in 1,625
HYAL1	?Mucopolysaccharidosis, type 9	1 in 2,811	N/A
HYLS1	Hydrolethalus syndrome	1 in 522	1 in 587
IDH3B	Retinitis pigmentosa, type 46	1 in 500	1 in 999
IDS	Mucopolysaccharidosis, type 2	1 in 38,000	1 in 90,827
IDUA	Mucopolysaccharidosis type 1	1 in 153	1 in 340
IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S	<1 in 500	<1 in 4,000
IL1RAPL1	Mental retardation, X-linked, type 21/34	1 in 45,000	1 in 90,000
IL2RG	Severe combined immunodeficiency, X-linked	1 in 33,334	<1 in 84,000
IVD	Isovaleric acidemia	1 in 115	1 in 1,749
IYD	Thyroid dyshormonogenesis, type 4	1 in 159	1 in 633
JAK3	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	1 in 475	1 in 732
KCNJ11	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	1 in 232	1 in 478
KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type	1 in 500,000	1 in 1,000,000
L1CAM	L1 Syndrome	1 in 15,000	<1 in 49,000
LAMA2	LAMA2-related muscular dystrophy	1 in 125	1 in 656
LAMA3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	1 in 606	1 in 60,501
LAMB3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	1 in 222	1 in 22,101
LAMC2	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	1 in 772	1 in 5,949
LCA5	Leber congenital amaurosis, type 5	1 in 1,811	1 in 2,237
LHCGR	Leydig cell hypoplasia	<1 in 500	<1 in 1,060
LHX3	Pituitary hormone deficiency, combined, type 3	1 in 1,398	1 in 9,780
LIFR	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	1 in 848	1 in 2,881
LIPA	Lysosomal acid lipase deficiency	1 in 112	1 in 176
LIPH	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	1 in 500	1 in 2,995
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	1 in 414	1 in 552
LOXHD1	Deafness, autosomal recessive, type 77	1 in 150	1 in 293
LPL	Lipoprotein lipase deficiency	1 in 374	1 in 1,120

LRP2	Donnai-Barrow syndrome	1 in 1000	1 in 50,000
LRPPRC	Leigh syndrome, French-Canadian type	1 in 768	1 in 1,560
LYST	Chediak-Higashi syndrome	N/A	N/A
MAN2B1	Alpha-mannosidosis	1 in 274	1 in 1,247
MANBA	Mannosidosis, beta	1 in 684	1 in 1,709
MAT1A	Methionine adenosyltransferase deficiency, autosomal recessive	1 in 708	1 in 1,920
MCCC1	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	1 in 353	1 in 705
MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	1 in 204	1 in 788
MCEE	Methylmalonyl-CoA epimerase deficiency	1 in 500	1 in 50,000
MCOLN1	Mucolipidosis type 4	1 in 1,166	1 in 4,850
MCPH1	Microcephaly type 1, primary, autosomal recessive	1 in 500	1 in 8,333
MECP2	Encephalopathy, neonatal severe; Rett syndrome	1 in 500,000	1 in 1,000,000
MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	1 in 1,287	N/A
MEFV	Familial Mediterranean fever	1 in 40	1 in 95
MESP2	Spondylocostal dysostosis, type 2, autosomal recessive	1 in 3,820	1 in 4,457
MFSD8	Ceroid lipofuscinosis, neuronal, type 7	1 in 555	1 in 1,272
MID1	Opitz GBBB syndrome, type 1	1 in 137	1 in 913
MKS1	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	1 in 246	1 in 859
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	1 in 884	1 in 4,416
MLYCD	Malonyl-CoA decarboxylase deficiency	1 in 500	1 in 1,000
MMAA	Methylmalonic aciduria, vitamin B12-responsive	1 in 677	1 in 2,619
MMAB	Methylmalonic aciduria, vitamin B12-responsive, type cblB	1 in 672	1 in 3,220
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	1 in 224	1 in 2,232
MMADHC	Homocystinuria, cblD type, variant 1	1 in 2,194	1 in 6,215
MOGS	Congenital disorder of glycosylation, type 2B	<1 in 500	<1 in 667
MPI	Congenital disorder of glycosylation, type 1B	1 in 473	1 in 9441
MPL	Thrombocytopenia, congenital amegakaryocytic	1 in 241	1 in 433
MPV17	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	1 in 612	1 in 1,783
MTHFR	Homocystinuria due to MTHFR deficiency	1 in 1,119	N/A
MTM1	Myotubular myopathy, X-linked	1 in 29,412	<1 in 140,000
MTMR2	Charcot-Marie-Tooth disease, type 4B1	1 in 500	1 in 1,000
MTR	Homocystinuria-megaloblastic anemia, cblG complementation type	<1 in 500	<1 in 864
MTRR	Homocystinuria-megaloblastic anemia, cbl E type	1 in 642	1 in 875
MTTP	Abetalipoproteinemia	1 in 655	1 in 1,496
MVK	Mevalonic aciduria	1 in 286	1 in 2,261
MYO15A	Deafness, autosomal recessive, type 3	N/A	N/A
MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	1 in 129	1 in 371
NADK2	2,4-dienoyl-CoA reductase deficiency	1 in 500	N/A
NAGA	Schindler disease, type I	1 in 500	1 in 5000
NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo B)	1 in 346	1 in 644
NAGS	N-acetylglutamate synthase deficiency	1 in 920	1 in 1,755
NBN	Nijmegen breakage syndrome	1 in 525	1 in 1,667
NCF1	Chronic granulomatous disease, type 1	1 in 343	1 in 1,027

NCF2	Chronic granulomatous disease, type 2	1 in 1,883	1 in 3,953
NDP	Norrie disease	1 in 50,000	<1 in 1,000,000
NDRG1	Charcot-Marie-Tooth disease, type 4D	1 in 7,299	1 in 29,193
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10	1 in 1,014	1 in 2,365
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16	1 in 982	1 in 1,262
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1	1 in 1,738	1 in 5,212
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9	1 in 3,535	1 in 4,419
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3	1 in 839	1 in 1,049
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	1 in 469	1 in 736
NEB	Nemaline myopathy type 2	1 in 175	1 in 517
NEU1	Sialidosis, type 1 and type 2	1 in 1,666	1 in 2,841
NHP2	Dyskeratosis congenita, autosomal recessive type 2	1 in 250	1 in 24,964
NLRP7	Hydatidiform mole, recurrent, type 1	1 in 500	1 in 722
NOP10	Dyskeratosis congenita, autosomal recessive type 1	1 in 250	1 in 500
NPC1	Niemann-Pick disease, type C1	1 in 163	1 in 954
NPC2	Niemann-pick disease, type C2	1 in 945	1 in 3,588
NPHP1	Joubert syndrome type 4	1 in 418	1 in 825
NPHS1	Nephrotic syndrome, type 1	1 in 190	1 in 1,993
NPHS2	Nephrotic syndrome, type 2	1 in 226	1 in 601
NR0B1	Adrenal hypoplasia, congenital	1 in 35,000	<1 in 66,000
NR2E3	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	1 in 278	1 in 417
NTRK1	Insensitivity to pain, congenital, with anhidrosis	1 in 1,122	1 in 1,974
OAT	Gyrate atrophy of choroid and retina	1 in 749	1 in 5,071
OCA2	Oculocutaneous albinism type 2	1 in 101	1 in 204
OCRL	Lowe Syndrome; Dent disease type 2	1 in 357,144	<1 in 1,000,000
OPA3	3-methylglutaconic aciduria, type 3	1 in 4,808	1 in 6,010
OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	1 in 500,000	1 in 1,000,000
OSTM1	Osteopetrosis, autosomal recessive type 5	1 in 6,173	1 in 9,259
OTC	Ornithine transcarbamylase deficiency	1 in 37,667	<1 in 96,000
OTOF	Deafness, autosomal recessive, type 9	1 in 228	1 in 22,701
PAH	Phenylketonuria	1 in 51	1 in 1,239
PAK3	Mental retardation, X-linked, type 30	1 in 45,000	1 in 90,000
PANK2	Neurodegeneration with brain iron accumulation type 1	1 in 700	1 in 1,175
PC	Pyruvate carboxylase deficiency	1 in 251	1 in 636
PCBD1	Hyperphenylalaninemia, BH4-deficient, type D	1 in 984	1 in 1,312
PCCA	Propionic acidemia	1 in 636	1 in 1,237
PCCB	Propionic acidemia	1 in 635	1 in 1,816
PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	1 in 497	1 in 1,034
PDE6A	Retinitis pigmentosa type 43	1 in 500	1 in 863
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	N/A	N/A
PDHB	Pyruvate dehydrogenase E1-beta deficiency	1 in 2,529	1 in 4,214
PEX1	Heimler syndrome type 1	1 in 191	1 in 19,000

PEX10	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B	1 in 1,287	1 in 4,245
PEX12	Peroxisome biogenesis disorder type 3A (Zellweger)	1 in 718	1 in 71,701
PEX2	Peroxisome biogenesis disorder type 5A (Zellweger)	1 in 1,542	1 in 6,165
PEX26	Peroxisome biogenesis disorder type 7A (Zellweger)	1 in 996	1 in 1,659
PEX5	Peroxisome biogenesis disorder type 2A (Zellweger)	1 in 914	1 in 2,010
PEX6	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	1 in 83	1 in 216
PEX7	Rhizomelic chondrodysplasia punctata, type 1	1 in 371	1 in 1,032
PFKM	Glycogen storage disease, type 7	1 in 868	1 in 1,843
PGK1	Phosphoglycerate kinase 1 deficiency	1 in 500,000	1 in 1,000,000
PHF8	Mental retardation syndrome, X-linked, Siderius type	1 in 500,000	1 in 916,000
PHGDH	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	1 in 631	1 in 3,361
PKHD1	Polycystic kidney disease type 4	1 in 66	1 in 266
PLA2G6	Infantile neuroaxonal dystrophy type 1	1 in 343	1 in 856
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	1 in 159	1 in 299
PLP1	Pelizaeus-Merzbacher disease	1 in 91,838	1 in 113,109
PMM2	Congenital disorder of glycosylation, type 1A	1 in 71	1 in 7,022
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	1 in 1,107	1 in 3,983
POLG	POLG-related disorders	1 in 194	1 in 340
POLR1C	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	1 in 1,021	1 in 1,659
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 31,401
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
POMT2	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	1 in 833	1 in 7,281
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	1 in 268	1 in 1,069
POU1F1	Pituitary hormone deficiency, combined, type 1	1 in 32	1 in 126
POU3F4	Deafness, X-linked, type 2	1 in 556,112	<1 in 1,000,000
PPM1K	?Maple syrup urine disease, mild variant	N/A	N/A
PPT1	Ceroid lipofuscinosis, neuronal, type 1	1 in 488	1 in 2,165
PQBP1	Renpenning syndrome	1 in 50,000	1 in 107,142
PRDX1	Methylmalonic aciduria and homocystinuria, cblC type, digenic	N/A	N/A
PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2	1 in 308	1 in 538
PRODH	Hyperprolinemia, type 1	N/A	N/A
PROP1	Pituitary hormone deficiency, combined, type 2	1 in 84	1 in 8,299
PRPS1	PRPS1-related disoders	<1 in 50,000	<1 in 221,000
PSAP	Combined SAP deficiency	1 in 2,039	1 in 3,873
PTS	Hyperphenylalaninemia, BH4-deficient, type A	1 in 478	1 in 1,259
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia, type 1	1 in 4,496	1 in 5,722
PYGM	McArdle disease	1 in 206	1 in 641
QDPR	Hyperphenylalaninemia, BH4-deficient, type C	1 in 2,419	1 in 5,182

RAB23	Carpenter syndrome	1 in 673	1 in 1,681
RAG1	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	1 in 344	1 in 614
RAG2	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	1 in 1,925	1 in 3,721
RAPSN	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	1 in 165	1 in 312
RARS2	Pontocerebellar hypoplasia, type 6	1 in 269	1 in 365
RAX	Isolated microphthalmia, type 3	1 in 159	1 in 475
RDH12	Leber congenital amaurosis, type 13	1 in 456	1 in 696
RNASEH2B	Aicardi-Goutieres syndrome, type 2	1 in 440	1 in 7,333
RNASEH2C	Aicardi-Goutieres syndrome, type 3	1 in 1,525	1 in 3,557
RP2	Retinitis pigmentosa, type 2, X-linked	1 in 177,778	1 in 249,736
RPE65	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy	1 in 366	1 in 496
RPGR	Retinitis pigmentosa, type 3, X-linked; Cone-rod dystrophy, X-linked, 1	1 in 31,373	1 in 35,705
RPGRIP1L	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	1 in 319	1 in 860
RS1	Retinoschisis	1 in 25,000	1 in 44,241
RTEL1	Dyskeratosis congenita, autosomal recessive type 5	1 in 800	N/A
SACS	Spastic ataxia, Charlevoix-Saguenay, type	1 in 100	1 in 146
SAG	Oguchi disease, type 1	N/A	N/A
SAMHD1	Aicardi-Goutieres syndrome, type 5	1 in 610	1 in 1,147
SBDS	Shwachman-Diamond syndrome	1 in 224	1 in 804
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1	1 in 500	1 in 8,333
SEPSECS	Pontocerebellar hypoplasia, type 2D	1 in 656	1 in 743
SERPINA1	Alpha-1 antitrypsin deficiency	1 in 24	<1 in 469
SGCA	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	1 in 288	1 in 1,938
SGCB	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	1 in 628	1 in 2,330
SGCD	Limb-girdle muscular dystrophy, type 6 (LGMD R6)	1 in 3,766	1 in 11,296
SGCG	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	1 in 1,132	1 in 5,468
SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo A)	1 in 253	1 in 1,261
SH2D1A	Lymphoproliferative syndrome, X-linked, type 1	<1 in 500,000	<1 in 829,000
SH3TC2	Charcot-Marie-Tooth disease, type 4C	1 in 72	1 in 184
SLC12A3	Gitelman syndrome	1 in 73	1 in 97
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	1 in 1,515	1 in 151,401
SLC16A2	Allan-Herndon-Dudley syndrome	1 in 50,000	1 in 67,647
SLC17A5	Salla disease	1 in 328	1 in 32,701
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	1 in 500	1 in 888
SLC19A3	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)	1 in 232	1 in 1,785
SLC22A5	Carnitine deficiency, systemic primary	1 in 251	1 in 544
SLC25A13	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	1 in 619	1 in 1,426
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	1 in 1,283	1 in 4,820
SLC25A20	Carnitine-acylcarnitine translocase deficiency	1 in 1,988	1 in 7,949
SLC26A2	Achondrogenesis, type 1B (diastrophic dysplasia)	1 in 129	1 in 428
SLC26A3	Diarrhea 1, secretory chloride, congenital	N/A	N/A

SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome	1 in 88	1 in 403
SLC35A1	Congenital disorder of glycosylation, type 2F	<1 in 500	<1 in 1,500
SLC35A3	?Arthrogryposis, mental retardation, and seizures	1 in 4,537	1 in 6,805
SLC35C1	Congenital disorder of glycosylation, type 2C	N/A	N/A
SLC35D1	Schneckenbecken dysplasia	N/A	N/A
SLC37A4	Glycogen storage disease, type 1B	1 in 597	1 in 1,677
SLC39A4	Acrodermatitis enteropathica	1 in 316	1 in 335
SLC3A1	Cystinuria	1 in 42	1 in 84
SLC45A2	Albinism, oculocutaneous, type 4	<1 in 500	<1 in 1,600
SLC46A1	Folate malabsorption, hereditary	1 in 2,468	1 in 13,981
SLC4A11	Corneal endothelial dystrophy, autosomal recessive	1 in 806	1 in 1,921
SLC5A5	Thyroid dyshormonogenesis, type 1	1 in 72	1 in 96
SLC6A19	Hartnup disorder	1 in 87	1 in 124
SLC6A8	Cerebral creatine deficiency syndrome, type 1	N/A	N/A
SLC7A7	Lysinuric protein intolerance	1 in 522	1 in 1,490
SLC7A9	Cystinuria	1 in 42	1 in 48
SMARCAL1	Schimke immunoosseous dysplasia	1 in 451	1 in 970
SMN1	Spinal muscular atrophy	1 in 36	1 in 869
SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B	1 in 350	1 in 1,556
SPG11	Amyotrophic lateral sclerosis, type 5, juvenile	1 in 192	1 in 467
SPG7	Spastic paraplegia, type 7, autosomal recessive	1 in 80	1 in 183
SRD5A2	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)	N/A	N/A
ST3GAL5	Salt and pepper developmental regression syndrome	N/A	N/A
STAR	Lipoid adrenal hyperplasia	1 in 1,147	1 in 14,326
SUMF1	Multiple sulfatase deficiency	1 in 696	1 in 2,665
SURF1	Charcot-Marie-Tooth disease, type 4K; Leigh syndrome, due to COX IV deficiency	1 in 191	1 in 329
SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	1 in 45,000	1 in 55,588
TAT	Tyrosinemia, type 2	1 in 2,190	1 in 5,352
TCIRG1	Osteopetrosis, autosomal recessive, type 1	1 in 399	1 in 1,195
TECPR2	Spastic paraplegia, type 49, autosomal recessive	1 in 1,946	N/A
TF	Atransferrinemia	1 in 500	1 in 7,143
TFR2	Hemochromatosis, type 3	1 in 604	1 in 1,885
TG	Thyroid dyshormonogenesis, type 3	1 in 159	1 in 268
TGM1	Ichthyosis, congenital, autosomal recessive, type 1	1 in 186	1 in 758
TH	Segawa syndrome, recessive	1 in 856	1 in 2,566
THOC2	Mental retardation, X-linked 12	1 in 500,000	N/A
TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2	1 in 1,521	1 in 152,001
TMEM67	Joubert syndrome, type 6; Meckel syndrome, type 3; COACH syndrome	1 in 147	1 in 284
TMPRSS3	Deafness, autosomal recessive, type 8/10	N/A	N/A
TNXB	Ehlers-Danlos syndrome, classic-like	1 in 50	1 in 250
TPO	Thyroid dyshormonogenesis, type 2A	1 in 72	1 in 84
TPP1	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	1 in 266	1 in 1,591

TRDN	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	1 in 51	1 in 71
TREX1	Aicardi-Goutieres syndrome, type 1	1 in 98	1 in 186
TRIM32	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	1 in 226	1 in 376
TRIM37	Mulibrey nanism	N/A	N/A
TRMU	Liver failure, transient infantile	1 in 789	1 in 1,001
TSEN54	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	1 in 223	1 in 3,997
TSFM	Combined oxidative phosphorylation deficiency, type 3	1 in 535	1 in 611
TSHB	Hypothyroidism, congenital, nongoitrous, type 4	1 in 62	1 in 306
TSHR	Hypothyroidism, congenital, nongoitrous, type 1	1 in 62	1 in 189
TTC37	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	1 in 634	1 in 951
TTPA	Ataxia with isolated vitamin E deficiency	1 in 607	1 in 3,637
TYMP	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	1 in 425	1 in 1,014
TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	1 in 101	1 in 681
TYRP1	Albinism, oculocutaneous, type 3	<1 in 500	<1 in 1,400
UBE3A	Angelman syndrome	1 in 62	N/A
UGT1A1	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	1 in 500	1 in 5,496
UNC13D	Hemophagocytic lymphohistiocytosis, familial, type 3	1 in 108	1 in 202
UPF3B	Mental retardation, X-linked, syndromic, type 14	1 in 45,000	1 in 49,500
USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	1 in 257	1 in 902
USH1G	Usher syndrome, type 1G	1 in 4,124	1 in 9,242
USH2A	Usher syndrome, type 2A	1 in 60	1 in 173
VPS13A	Choreoacanthocytosis	1 in 341	1 in 488
VPS13B	Cohen syndrome	1 in 224	1 in 610
VPS45	Neutropenia, severe congenital, type 5	1 in 1,634	N/A
VPS53	Pontocerebellar hypoplasia, type 2E	1 in 1,506	N/A
VRK1	Pontocerebellar hypoplasia, type 1A	1 in 2,583	1 in 3,377
VSX2	Microphthalmia with coloboma 3; Isolated microphthalmia 2	1 in 1,337	1 in 3,564
WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	1 in 500,000	<1 in 1,000,000
WNT10A	Odontoonychodermal dysplasia	1 in 216	1 in 324
WRN	Werner syndrome	1 in 474	1 in 681
XPA	Xeroderma pigmentosum, group A	1 in 718	1 in 4,877
XPC	Xeroderma pigmentosum, group C	1 in 440	1 in 1,635
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type	1 in 45,000	1 in 60,000
ZFYVE26	Spastic paraplegia, type 15, autosomal recessive	<1 in 500	<1 in 1,600
ZNF711	Mental retardation, X-linked, type 97	1 in 45,000	1 in 54,000