CGT Exome v5.4.5

Patient Information		Sample Information		Clinic Information	
Unique pat id.:	0246001	**Sample type:**	Blood	**Clinic:**	WeFIV
Patient name:		**Date of draw:**	13/06/2024	**Doctor:**	FERNANDO NEUSPILLER
Patient DOB:		**Date of receipt:**	18/06/2024		
Ethnic group:	Hispanic	**Report date/time:**	14/04/2026		

Indication: No family history

TEST RESULTS

NEGATIVE

INTERPRETATION OF TEST RESULTS

For a negative test result, the risk of having children affected by the investigated conditions decreases significantly compared to the general population. This is also the case for a negative personal result when a partner or 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)

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

The report language has been changed at the clinic’s request.

TEST METHODOLOGY

DNA is isolated from the sample, usually blood or saliva, and analyzed by whole exome sequencing by NGS. This includes 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), which includes sequence alignment against the GRCh37 human genome reference, variant calling, annotation, and real-time interpretation of variants. QC parameters include, all reported samples that 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) are performed for the following genes, if included, CFTR gene intronic variant/s; SMN1 gene exon 7-deletion; CYP21A2 gene frequent mutations; HBA1 and HBA2 genes frequent deletions; FXN gene GAA repeat sizing; FMR1gene CGG repeat sizing (females only); DMD gene frequent deletions/duplications; F8 gene intron 22 inversion (females only). When requested, CNV analysis by MLPA is performed for CFTR, HBB and HBA1/HBA2. 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. The CYP21A2 gene analysis presents unique challenges due to its high sequence homology (~98%) with the pseudogene CYP21A1P, which leads to frequent gene rearrangements and complex mutations. These challenges can cause difficulties in distinguishing CYP21A2 from CYP21A1P, increasing the risk of misdiagnosis. Different testing methods have specific limitations, requiring a combination of techniques such as long-range PCR, Sanger sequencing, next-generation sequencing (NGS), MLPA, and qPCR to achieve accurate results. These challenges and limitations may lead to false or inconclusive results. Therefore, genetic counselling is strongly recommended to evaluate the findings, discuss potential implications, and determine whether additional testing (such as MLPA) is necessary for an accurate diagnosis. In summary, sensitivity to detect pathogenic variants, if they result from complex gene conversion/gene rearrangements events, may be reduced. For the HEXB gene, the common 16 kb deletion that causes disease in 30% of affected patients is not included in CGT analysis. Furthermore, this test does not evaluate the HFE gene.

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

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 SPAIN LAB, SLU. 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.

Part of this test has been outsourced to a reference laboratory whose Quality Management System is based on high Quality Standards, periodically monitored by Igenomix SPAIN* and audited by independent external groups.

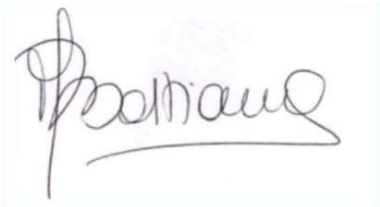
*IGENOMIX SPAIN holds CLIA Certificate of Compliance: #99D2146167.

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	COUNTERSIGNED
	
Martina Di Bastiano	**Arantxa Hervas PhD**
Laboratory Leader	3025-CV Biotechnologist

This test or part of this test has been outsourced to a referral Laboratory. Lab CLIA No.: 99D2146167

LIST OF ANALYZED GENES

Gene mean coverage >100x CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, ACO2, APRT, ACTA1, ADK, AMPD1, AMPD2, SLC25A4, FDXR, AGRN, ALB, ALDOA, AFP, NAGA, SLC3A1, ACY1, AGT, AGTR1, ACE, CD19, SERPINC1, ALDH7A1, SERPINA1, IFNGR1, AQP2, RARS1, CYP19A1, DDC, ASNS, ATP2A1, ATP6V1E1, NPR2, SLC4A1, CD40, B2M, BLVRA, CD79A, BMP1, DST, COL17A1, CAD, CDH3, CAST, CACNA1D, CAPN1, CAPN3, CASQ2, CA5A, CAT, TYRP1, CTSD, CP, HSPD1, CLCN1, CYP11A1, COL4A3, COL7A1, COL4A4, COL1A2, COL9A1, COL6A1, COL6A2, COL6A3, COL9A2, COL11A1, COL11A2, COL18A1, COL13A1, MMP2, DCC, C1S, CR2, C3, C5, C8B, GJB2, GJA1, CRYAA, CRYAB, CRYBB3, CNGA1, PPIB, CTPS1, IL10RB, CYC1, POR, CYP11B2, COX6B1, CD55, ALAD, DSP, DES, DSG1, CYP24A1, PCBD1, ERCC2, SLC26A3, EGR2, NT5E, ETFB, TYMP, EDN3, EDNRB, ENO3, ERCC3, ERCC4, ERCC5, SLC1A1, CFH, F13A1, F13B, FTL, FGA, FGB, FGG, FLG, FMO3, FOLR1, FSHR, FSHB, FH, ABAT, GGCX, TACSTD2, MPV17, GCK, SLC2A1, GOT2, SLC2A2, GRIK2, GRIN1, ALDH18A1, GFPT1, EPRS1, GPX4, GLRA1, GLRB, GYS1, GYS2, GNRHR, GRN, CSF3R, GHRHR, GH1, GNAT1, GNAT2, HBA1, HBA2, HBB, HGF, HSPG2, HK1, HARS1, HOXA1, VSX2, HADHB, IL7R, IGLL1, ADAR, CD79B, ITPR1, IGF1R, IGF1, SOD1, ITGA6, ITGB4, ITGB6, ISG15, INSR, IL1RN, IL2RA, KRT5, KRT14, KRT10, LDHA, LAMB1, LAMC2, LAMB3, LAMA1, LAMB2, LARS1, LIFR, PTPRC, LIPE, LHB, LHCGR, LCK, PLOD1, LIM2, MAK, MGP, LAMA2, MARS1, MTR, MITF, MAPT, MTTP, MPZ, MAG, MPL, MYH2, MYO5A, MYBPC1, NDUFV1, IL12B, NEB, NGF, PCSK1, NEFL, TAC3, PNP, LEP, SPINK1, PTH, PTH1R, TAP1, PRF1, PEX2, ABCB4, ALPL, PGM1, GPI, MTHFD1, PHKG2, PSPH, PHKB, SERPINF1, POU1F1, JUP, ENPP1, PLG, SERPINE1, ITGB3, POLE, POLG, KCNE1, INS, PRDX1, PSAP, PROS1, F2, TYK2, ZAP70, PRKCD, PSMB8, EPB42, BCHE, PYCR1, PDHB, RDX, PRPH2, RAG1, RAG2, REN, RD3, RPE65, PDE6A, PDE6B, RLBP1, RARB, RBP4, RBP3, RHO, ROM1, RYR1, AHCY, SAG, STIL, SPR, SLC34A1, SLC5A1, SLC5A2, SPTA1, CSTA, EPCAM, SURF1, CD27, CD3D, CD3E, ITK, TERT, TK2, TG, TSHB, TTN, TFRC, ERBB3, THRB, TGM1, SLC25A1, TNNT1, TH, NTRK1, UQCRC2, UCHL1, DPAGT1, UNG, UGT1A1, ATP6V1B1, VARS1, XRCC4, ZP1, BBS1, CFI, C6, C7, KLKB1, ETFDH, PCCA, PCCB, GLDC, AMT, GCSH, DLD, INVS, DBT, BCKDHB, MVK, IDUA, F11, TBXAS1, MYO7A, ATP7A, ATRX, DKC1, ARSL, IL1RAPL1, HSD17B10, UPF3B, BTK, CUL4B, DMD, EMD, CD40LG, WAS, THOC2, MTM1, OTC, PQBP1, CYBB, SH2D1A, PDHA1, OCRL, FGD1, BRWD3, PHF8, GLA, F9, RP2, GPR143, F8, COL4A5, G6PD, HPRT1, IL2RG, L1CAM, FMR1, PRPS1, RPGR, SYN1, KDM5C, ZNF711, LBR, ABCA1, CNGA3, ITGB2, LRP2, MMP13, SGCA, JAK3, GUCY2D, GCH1, SCNN1A, HMGCS2, CENPF, PEX19, PI4KA, ACADSB, KCNJ1, BBS4, PEX5, TTPA, NT5C2, GCNT2, SKIC2, IGHMBP2, ABCC8, RELN, AUH, NDUFV2, ITGA7, HSPA9, STAT1, STAT2, NRXN1, CLCN2, LZTR1, PTPRO, STAR, CPT2, CHUK, SLC16A1, IARS1, PPT1, CNGB1, SCNN1B, SCNN1G, TAF13, LAMA3, PLCB4, DDB2, MRE11, PDE6C, INPPL1, SLC12A1, SDHA, AIMP2, MSH3, HADHA, SGCB, CRYBB1, IL11RA, SERPINH1, DHPS, GHR, AMHR2, SLC12A3, MYO6, DMP1, GSS, NPC2, AARS1, PMP22, CTSK, ABCC2, CSTB, GDF6, DDX11, PDE6H, ATR, BIN1, KIF1A, AMBN, C1QBP, PLEC, HINT1, GUCY2C, MOGS, TUSC3, 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PGAM2, TACO1, PDZD7, TMEM126A, RAB28, SEPSECS, TAT, PMPCA, INPP5E, LOXHD1, GM2A, CTSA, RETREG1, CTC1, VWF, CANT1, BOLA3, DNAAF1, XPC, WDR72, AGA, PEPD, TMEM216, SLX4, GRXCR1, SH3PXD2B, DPYS, OAT, DRAM2, DYNC2I2, CBS, POMP, AVIL, VIPAS39, PCARE, TCN2, CEP120, ASAH1, LIPA, UROD, SDCCAG8, CEP152, FAN1, XPNPEP3, TBC1D24, WDR62, FAM161A, HOGA1, ABHD12, WDR35, NUBPL, FOXRED1, PIEZO2, AP5Z1, SLC25A20, ANO10, PYGL, G6PC1, CCDC39, CCDC40, TTC19, CYP21A2, TCTN2, TCTN3, PRSS56, FAH, F10, F7, HSD3B2, UMPS, DPY19L2, FANCF, HMGCL, FANCC, LIPN, C2, TOE1, SUN5, FANCE, FANCD2, BPNT2, IFT43, WEE2, PCK2, TRAPPC11, TRAPPC12, PRDM5, NBEAL2, DIS3L2, DOCK6, POLR3A, PAM16, POLR3B, MFSD2A, TMEM237, TTI2, TMEM138, GPR179, NDUFA12, DNAAF3, CPLANE1, ROGDI, SKIC3, IFT140, CRPPA, COQ6, PATL2, CCDC103, SERAC1, TMEM165, PIGO, CFAP53, PET100, POC1A, POC1B, MFF, EOGT, CEP164, DNAAF5, BCKDK, HIKESHI, RMND1, OTOGL, DNAAF11, EPS8L2, LRIT3, ODAD1, EPG5, MGME1, C12orf57, PGAP2, DRC1, GMPPB, KLHL40, MYMK, THOC6, ODAD2, TRMT10C, DYNC2I1, SZT2, DDX59, GMPPA, STAC3, TMEM126B, SYNE4, SFXN4, SLC38A8, KPTN, HFM1, DOCK7, WASHC4, KIZ, CEP83, AGBL5, C2CD3, SPEG, ODAD3, COA8, SNX14, CWF19L1, TGDS, CKAP2L, DDRGK1, ZNF408, RSPRY1, KRT25, KATNIP, CEP104, CCDC174, MAPKBP1, EMC1, PIGG, GUF1, DYNC2LI1, IFT52, KIAA0753, CWC27, TMTC3, NSMCE2, DENND5A, DYNLT2B, TMEM260, EFL1, CFAP43, DZIP1L, TBC1D23

Gene mean coverage 50x-100x AK1, AK2, APOE, BCAT2, CALCRL, CHAT, MME, C1QA, C1QB, C1QC, DHFR, DHODH, ERCC1, SLC6A3, DAG1, EPB41, EDN1, EGFR, CFD, B4GALT1, GPT2, GLUL, GPD1, GSC, CSF2RB, HMOX1, HPCA, IL10RA, ITPA, IFNGR2, MDH2, MPI, NDUFS1, RMRP, TACR3, CHMP1A, FGF3, TAP2, ACP5, PGM3, CD36, GP9, POMC, SFTPB, PDE6G, GRK1, SPARC, SLC9A3, CD3G, CD247, CD81, TRHR, TPI1, TPM3, DDR2, VLDLR, FLI1, HCFC1, SLC6A8, SLC16A2, OPHN1, DLG3, ABCD1, ARX, CHM, PLP1, NR0B1, FTSJ1, MID1, ZDHHC9, NDP, RS1, GJB1, PGK1, CDH11, SLC1A4, SLC18A3, SLC25A3, AKR1C2, ALDH1A3, P2RY12, MASP1, SLC11A2, CPT1A, LHX3, ASPH, NECTIN1, IHH, CEBPE, FOXN1, NDST1, LSS, KCNJ11, AMH, TNXB, LEPR, SLC6A9, CLPP, GDF5, CASR, GAMT, TOP3A, SLC10A2, MUSK, SLCO2A1, MYO1E, IRF8, UBE3A, SERPINB8, HESX1, NDUFS7, B4GALNT1, RAX, WNT10B, ARHGDIA, TAPBP, IMPA1, ESRRB, MYD88, ECM1, SCARB2, LAT, WIPF1, CTSC, FUT8, CDSN, PEX10, ORC4, KIF1C, CAVIN1, TRDN, CRADD, TNFRSF11A, BCL10, DPH1, AP1S1, CTSF, DPM2, AIMP1, COX15, CYP7B1, SGPL1, MED17, NDUFA10, PEX11B, GOSR2, GRM6, TBX15, DNAJB2, SLC6A5, KISS1R, STAT5B, AGXT, BLOC1S6, B4GALT7, LAMC3, ST3GAL5, PLPBP, SP110, GRM1, ZNHIT3, BLNK, ZNF423, WARS2, MALT1, MKKS, DGAT1, BRF1, STK4, SPTBN2, CORO1A, STX11, SPINT2, GAS8, MESP2, CHST6, IL21R, ALX4, ABCG8, B4GAT1, CDT1, ATF6, CIB2, CLDN14, PCDH12, PRX, SOST, DCDC2, NUP62, TMC6, TMC8, SLC35C1, ALG1, STIM1, DPM3, DCLRE1C, MBOAT7, PIP5K1C, SLC19A3, IRX5, SLC45A2, SIX6, ESPN, ANTXR1, STRC, EIF2B2, NHP2, CD320, LZTFL1, SFRP4, OPA3, SP7, UPB1, HEXA, JAM3, COG8, ATP6V1A, LIAS, CLN3, HYAL1, MTHFR, MCPH1, ALG12, FYCO1, PTF1A, TMIE, SRD5A2, AIRE, LIPH, DYM, HPS6, EPM2A, ARSA, ADA2, COLEC10, CCNO, HSD3B7, MCFD2, GNPTG, RCBTB1, SLC52A2, ASPA, NHLRC1, CLN5, PUS1, XYLT2, TTC8, CHSY1, ADSL, NEU1, LGI4, CHST14, SMOC1, NCF1, GTPBP3, GLIS2, GRHL2, GLDN, PTRH2, PSMC3IP, SLC39A8, SLC39A13, SLC39A14, TSEN15, SLC5A7, GJC2, ARL6, LRIG2, SLC6A19, ARL13B, CABP4, MRPS16, WDR45B, LPAR6, SLC25A22, TTC7A, REEP6, ERCC8, ERCC6, MAP3K20, NECTIN4, NAGLU, CRB2, MARS2, NEK9, MMACHC, RNF216, COL25A1, CLDN19, DNAL1, PRMT7, MCM9, B3GALNT2, MED25, PIGV, PIGW, ORAI1, DOK7, SLC6A17, LINS1, TRAPPC6B, GLYCTK, HYCC1, UBA5, UFM1, PDSS2, MARVELD2, FLAD1, PRCD, GINS1, PDE10A, PIGY, CYP2U1, HYLS1, CNPY3, HYDIN, SLC25A46, RBCK1, FTO, OBSL1, TRMT5, SLC25A26, RUSC2, FAM20C, FAM20A, SLC30A10, FARS2, ERLIN1, ARV1, PGAP1, TBC1D20, LMF1, MTFMT, NKX2-6, PGAP3, ISCU, B9D2, TRAPPC9, MRPS34, OTUD6B, IYD, ZNF469, PNPLA1, ADAMTSL2, CILK1, NDUFAF5, FECH, RSPH9, TBC1D7, DSTYK, RNF168, GPIHBP1, SDHAF1, COQ4, TRNT1, NDUFAF3, SERPINF2, SMG9, CYB5R3, PPP1R15B, MOCOS, USB1, ZC3H14, FEZF1, KIAA1549, SLC52A3, TPRN, HPSE2, MTRFR, SCARF2, ALG11, SOBP, GZF1, BPGM, AFG2A, ZBTB24, MCIDAS, TRIM2, CCDC8, WDR81, HSD11B2, ACSF3, C19orf12, NUP93, ATAD1, CFAP418, BRAT1, LRMDA, KANK2, UVSSA, MPC1, COA6, SNX10, POMGNT2, DHTKD1, TECPR2, DDHD2, POMK, METTL23, CERS3, ADAT3, ANKS6, ARL2BP, TIMMDC1, COQ8B, CEP19, CDIN1, ERCC6L2, NADK2, VPS53, JAGN1, TRMT10A, ELP2, SPRTN, FAR1, LMOD3, TMEM107, LEMD2, PYCR2, ARHGEF18, PRDM12, TMEM199, TANGO2, TBCK, DNAJC21, PYROXD1, TECRL, PRUNE1, CLDN10, ALPK3, ARMC9, MSTO1, IQCE, TRIT1

Gene mean coverage < 50x LRPAP1, CD59, CA8, PCYT1A, ESR1, KDSR, H6PD, GP1BB, HOXB1, HOXC13, HMX1, NRL, WNT1, WNT3, PAX7, PRRX1, ZBTB16, PDXK, CD8A, TF, UQCRB, MECP2, POU3F4, DCX, PAK3, EDA, AP1S2, IDS, AR, CIITA, MEOX1, SCN1B, SMN1, PDX1, GFER, FOXE3, GPR68, DGKE, WNT7A, SOX18, ENTPD1, NKX3-2, EMP2, FADD, FOXE1, PHOX2A, OCLN, CA12, SCO1, SLC33A1, CLDN1, NDUFA9, GYG1, RNASEH1, GNB5, FRRS1L, CRIPT, NEUROG3, CPLX1, S1PR2, KYNU, CYP26B1, RBM8A, GAN, DHH, SLC35A3, FBXL4, KY, EXTL3, NKX6-2, ALX3, PANK2, STAMBP, BSND, MPIG6B, FKRP, MLYCD, FXN, AP4S1, TIMM50, TWIST2, CLCF1, SAR1B, CNNM2, CLN8, HES7, XYLT1, CYP26C1, PEX26, TP53RK, NMNAT1, GTF2H5, BTD, MPLKIP, IER3IP1, L2HGDH, GLRX5, BLOC1S3, SLC24A4, ATOH7, KREMEN1, VPS37A, PIGM, DCPS, RSPO4, SLC35D1, SYT14, ISCA1, PPM1K, NHEJ1, CISD2, TXNL4A, ERLIN2, CLMP, NDUFAF6, NDUFA11, RNASET2, CYB5A, POLR1D, MSRB3, CCDC115, TMCO1, B9D1, FKBP14, DDHD1, MTO1, COX20, TMEM231, B3GALT6, IBA57, ISCA2, BHLHA9, OTULIN, SLC7A14, LYRM7, RSPH3, WDR73, CLPB, CEP78, LIPT2

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	ABCD1	Adrenoleukodystrophy	1 in 3750	1 in 37500
X	AP1S2	Mental retardation, X-linked, syndromic, type 5 (Pettigrew syndrome)	< 1 in 100 000	Reduced
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	ATRX	Mental retardation-hypotonic facies syndrome, X-linked; Alpha-thalassemia/mental retardation syndrome	< 1 in 100 000	Reduced
X	BRWD3	Mental retardation, X-linked, type 93	1 in 10000	1 in 50000
X	BTK	Agammaglobulinemia X-linked, type 1	1 in 50000	1 in 333333
X	CD40LG	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	< 1 in 100 000	Reduced
X	CHM	Choroideremia	1 in 18750	1 in 66964
X	COL4A5	Alport syndrome, X-linked	1 in 10000	1 in 50000
X	CUL4B	Mental retardation, X-linked, syndromic, type 15 (Cabezas type)	< 1 in 100 000	Reduced
X	CYBB	Chronic granulomatous disease, X-linked	1 in 300	1 in 1500
X	DCX	Lissencephaly, X-linked, type 1	1 in 2500	1 in 50000
X	DKC1	Dyskeratosis congenita, X-linked	1 in 62500	1 in 1250000
X	DLG3	Mental retardation, X-linked, type 90	1 in 45000	1 in 300000
X	DMD	DMD-related conditions	1 in 2625	1 in 131250
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 3500	1 in 89285
X	F9	Hemophilia B	1 in 6250	1 in 62500
X	FGD1	Aarskog-Scott syndrome; Mental retardation, X-linked syndromic, type 16	1 in 10000	1 in 125000
X	FMR1	FMR1-related conditions	1 in 400	1 in 40000
X	FTSJ1	Mental retardation, X-linked 44	1 in 45000	1 in 300000
X	G6PD	G6PD deficiency	1 in 25	1 in 250
X	GJB1	Charcot-Marie-Tooth neuropathy, X-linked dominant, type 1	1 in 9803	1 in 196060
X	GLA	Fabry disease	1 in 18750	1 in 187500
X	GPR143	Ocular albinism, type 1 (Nettleship-Falls type)	1 in 15000	1 in 18750
X	HCFC1	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	< 1 in 100 000	Reduced
X	HPRT1	Lesch-Nyhan syndrome	1 in 95000	1 in 380000
X	HSD17B10	HSD10 mitochondrial disease	< 1 in 100 000	Reduced
X	IDS	Mucopolysaccharidosis, type 2	1 in 25000	1 in 125000
X	IL1RAPL1	Mental retardation, X-linked, type 21/34	1 in 25000	1 in 357143
X	IL2RG	Severe combined immunodeficiency, X-linked	1 in 25000	1 in 500000
X	KDM5C	Mental retardation, X-linked, syndromic, Claes-Jensen type	1 in 4000	1 in 57143
X	L1CAM	L1 Syndrome	1 in 7500	1 in 150000
X	MECP2	Encephalopathy, neonatal severe; Rett syndrome	1 in 37500	1 in 250000
X	MID1	Opitz GBBB syndrome, type 1	1 in 18750	1 in 125000
X	MTM1	Myotubular myopathy, X-linked	1 in 12500	1 in 83333
X	NDP	Norrie disease	1 in 50000	< 1 in 1000000
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	OPHN1	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	< 1 in 500	Reduced
X	OTC	Ornithine transcarbamylase deficiency	1 in 50000	1 in 166667
X	PAK3	Mental retardation, X-linked, type 30	1 in 40000	1 in 800000
X	PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	< 1 in 100 000	Reduced
X	PGK1	Phosphoglycerate kinase 1 deficiency	< 1 in 100 000	Reduced
X	PHF8	Mental retardation syndrome, X-linked, Siderius type	< 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 556112	< 1 in 1000000
X	PQBP1	Renpenning syndrome	< 1 in 100 000	Reduced
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	RS1	Retinoschisis	1 in 15000	1 in 100000
X	SH2D1A	Lymphoproliferative syndrome, X-linked, type 1	< 1 in 100 000	Reduced
X	SLC16A2	Allan-Herndon-Dudley syndrome	< 1 in 100 000	Reduced
X	SLC6A8	Cerebral creatine deficiency syndrome, type 1	< 1 in 100 000	Reduced
X	SYN1	Epilepsy, X-linked, with variable learning disabilities and behavior disorders	1 in 30000	1 in 150000
X	THOC2	Mental retardation, X-linked 12	< 1 in 100 000	Reduced
X	UPF3B	Mental retardation, X-linked, syndromic, type 14	1 in 15000	1 in 75000
X	WAS	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked	< 1 in 100 000	Reduced
X	ZDHHC9	Intellectual developmental disorder, X-linked syndromic, Raymond type	1 in 45000	1 in 450000
X	ZNF711	Mental retardation, X-linked, type 97	1 in 45000	1 in 225000

Autosomal recessive conditions				
Chrom	**Gene**	**Disease/Condition**	**Carrier Rate**	**Residual Risk**
12	AAAS	Triple-A syndrome (achalasia-addisonianism-alacrimia)	1 in 436	1 in 8266
16	AARS1	Epileptic encephalopathy, early infantile, type 29	< 1 in 500	Reduced
6	AARS2	Combined oxidative phosphorylation deficiency 8; Leukoencephalopathy, progressive, with ovarian failure	< 1 in 500	Reduced
7	AASS	Hyperlysinemia, type 1 and type 2	< 1 in 500	Reduced
16	ABAT	GABA-transaminase deficiency	< 1 in 500	Reduced
9	ABCA1	Tangier disease	< 1 in 500	Reduced
2	ABCA12	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	1 in 194	1 in 715
16	ABCA3	Surfactant metabolism dysfunction, pulmonary, type 3	1 in 500	1 in 7143
1	ABCA4	Stargardt disease 1; Retinitis pigmentosa 19; Cone-rod dystrophy 3	1 in 62	1 in 3100
2	ABCB11	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	1 in 276	1 in 3450
7	ABCB4	Cholestasis, progressive familial intrahepatic, type 3	< 1 in 500	Reduced
10	ABCC2	Dubin-Johnson syndrome	< 1 in 500	Reduced
16	ABCC6	Pseudoxanthoma elasticum; Generalized arterial calcification of infancy, type 2	< 1 in 500	Reduced
11	ABCC8	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	1 in 192	1 in 1920
14	ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	1 in 496	1 in 49501
2	ABCG5	Sitosterolemia 2	< 1 in 500	Reduced
2	ABCG8	Sitosterolemia 1	< 1 in 500	Reduced
20	ABHD12	PHARC syndrome (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataract)	< 1 in 500	Reduced
3	ABHD5	Chanarin-Dorfman syndrome	< 1 in 500	Reduced
11	ACAD8	Isobutyryl-CoA dehydrogenase deficiency	< 1 in 500	Reduced
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 102	1 in 10200
10	ACADSB	Short/branched-chain acyl-CoA dehydrogenase deficiency	1 in 500	1 in 1125
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
22	ACO2	Infantile cerebellar-retinal degeneration	< 1 in 500	Reduced
17	ACOX1	Peroxisomal acyl-CoA oxidase deficiency	< 1 in 500	Reduced
3	ACOX2	Bile acid synthesis defect, congenital, type 6	< 1 in 500	Reduced
19	ACP5	Spondyloenchondrodysplasia with immune dysregulation	< 1 in 500	Reduced
16	ACSF3	Combined malonic and methylmalonic aciduria	1 in 90	1 in 900
1	ACTA1	Nemaline myopathy 3; Congenital fiber-type disproportion myopathy 1	< 1 in 500	Reduced
3	ACY1	Aminoacylase 1 deficiency	< 1 in 500	Reduced
20	ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	1 in 390	1 in 2600
22	ADA2	Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome; Sneddon syndrome	< 1 in 500	Reduced
8	ADAM9	Cone-rod dystrophy 9	< 1 in 500	Reduced
19	ADAMTS10	Weill-Marchesani syndrome, type 1, recessive	< 1 in 500	Reduced
9	ADAMTS13	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)	1 in 334	1 in 11100
15	ADAMTS17	Weill-Marchesani syndrome, type 4, recessive	< 1 in 500	Reduced
16	ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus	< 1 in 500	Reduced
5	ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	< 1 in 500	Reduced
9	ADAMTSL2	Geleophysic dysplasia type 1	< 1 in 500	Reduced
1	ADAMTSL4	Ectopia lentis et pupillae; Ectopia lentis, isolated, type 2	1 in 500	1 in 50000
1	ADAR	Aicardi-Goutieres syndrome, type 6	< 1 in 500	Reduced
19	ADAT3	Mental retardation, autosomal recessive 36	< 1 in 500	Reduced
16	ADGRG1	Polymicrogyria, bilateral frontoparietal	< 1 in 500	Reduced
6	ADGRG6	Lethal congenital contracture syndrome 9	< 1 in 500	Reduced
5	ADGRV1	Usher syndrome, type 2C	1 in 80	1 in 800
10	ADK	Hypermethioninemia due to adenosine kinase deficiency	1 in 500	1 in 1498
22	ADSL	Adenylosuccinase deficiency	< 1 in 500	Reduced
14	ADSS1	Myopathy, distal, 5	< 1 in 500	Reduced
4	AFG2A	Epilepsy, hearing loss, and mental retardation syndrome	< 1 in 500	Reduced
18	AFG3L2	Spastic ataxia, type 5, autosomal recessive	< 1 in 500	Reduced
4	AFP	Alpha-fetoprotein deficiency	< 1 in 500	Reduced
4	AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)	< 1 in 500	Reduced
2	AGBL5	Retinitis pigmentosa 75	< 1 in 500	Reduced
7	AGK	Cataract 38; Sengers syndrome	< 1 in 500	Reduced
1	AGL	Glycogen storage disease, type 3	1 in 200	1 in 2000
9	AGPAT2	Congenital generalized lipodystrophy (Berardinelli-Seip syndrome)	< 1 in 500	Reduced
2	AGPS	Rhizomelic chondrodysplasia punctata, type 3	< 1 in 500	Reduced
1	AGRN	Myasthenic syndrome, congenital, type 8	< 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
20	AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	< 1 in 500	Reduced
6	AHI1	Joubert syndrome, type 3	1 in 334	1 in 706

12	AICDA	Immunodeficiency with hyper-IgM, type 2	< 1 in 500	Reduced
4	AIMP1	Leukodystrophy, hypomyelinating, type 3	< 1 in 500	Reduced
7	AIMP2	Leukodystrophy, hypomyelinating, type 17	< 1 in 500	Reduced
17	AIPL1	Leber congenital amaurosis, type 4	1 in 400	1 in 1060
21	AIRE	Autoimmune polyendocrinopathy syndrome, type 1	1 in 310	1 in 4429
9	AK1	Hemolytic anemia due to adenylate kinase deficiency	< 1 in 500	Reduced
1	AK2	Reticular dysgenesis	< 1 in 500	Reduced
10	AKR1C2	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency	< 1 in 500	Reduced
7	AKR1D1	Bile acid synthesis defect, congenital, type 2	< 1 in 500	Reduced
9	ALAD	Porphyria, acute hepatic	< 1 in 500	Reduced
4	ALB	Analbuminemia	< 1 in 500	Reduced
10	ALDH18A1	Spastic paraplegia, type 9B, autosomal recessive; Cutis laxa, type 3A (De Barsy syndrome)	< 1 in 500	Reduced
15	ALDH1A3	Microphthalmia, isolated 8	< 1 in 500	Reduced
17	ALDH3A2	Sjogren-Larsson syndrome	< 1 in 500	Reduced
1	ALDH4A1	Hyperprolinemia, type 2	1 in 500	1 in 49951
6	ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	< 1 in 500	Reduced
14	ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency	< 1 in 500	Reduced
5	ALDH7A1	Epilepsy, pyridoxine-dependent	1 in 127	1 in 2540
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 1740
13	ALG11	Congenital disorder of glycosylation, type 1P	< 1 in 500	Reduced
22	ALG12	Congenital disorder of glycosylation, type 1G	< 1 in 500	Reduced
9	ALG2	Myasthenic syndrome, congenital, type 14, with tubular aggregates	< 1 in 500	Reduced
3	ALG3	Congenital disorder of glycosylation, type 1D	< 1 in 500	Reduced
1	ALG6	Congenital disorder of glycosylation, type 1C	1 in 500	1 in 5000
11	ALG8	Congenital disorder of glycosylation, type 1H	< 1 in 500	Reduced
11	ALG9	Congenital disorder of glycosylation, type 1L; Gillessen-Kaesbach-Nishimura syndrome	< 1 in 500	Reduced
2	ALMS1	Alström syndrome	1 in 250	1 in 1667
17	ALOX12B	Ichthyosis, congenital, autosomal recessive, type 2	< 1 in 500	Reduced
17	ALOXE3	Ichthyosis, congenital, autosomal recessive, type 3	1 in 200	1 in 1500
15	ALPK3	Cardiomyopathy, familial hypertrophic, type 27	< 1 in 500	Reduced
1	ALPL	ALPL-related conditions	1 in 274	1 in 2740
2	ALS2	Amyotrophic lateral sclerosis, type 2, juvenile; Primary lateral sclerosis, juvenile; Spastic paralysis, infantile onset ascending	< 1 in 500	Reduced
12	ALX1	Frontonasal dysplasia, type 3	< 1 in 500	Reduced
1	ALX3	Frontonasal dysplasia, type 1	< 1 in 500	Reduced
11	ALX4	Frontonasal dysplasia, type 2	< 1 in 500	Reduced
5	AMACR	Bile acid synthesis defect, congenital, type 4; Alpha-methylacyl-CoA racemase deficiency	< 1 in 500	Reduced
4	AMBN	Amelogenesis imperfecta, type IF	< 1 in 500	Reduced
19	AMH	Persistent Mullerian duct syndrome, type 1	< 1 in 500	Reduced
12	AMHR2	Persistent Mullerian duct syndrome, type II	< 1 in 500	Reduced
14	AMN	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)	< 1 in 500	Reduced
1	AMPD1	Myopathy due to myoadenylate deaminase deficiency	< 1 in 500	Reduced
1	AMPD2	Pontocerebellar hypoplasia, type 9	< 1 in 500	Reduced
3	AMT	Glycine encephalopathy	1 in 310	1 in 6200
1	ANGPTL3	Hypobetalipoproteinemia, familial, type 2	< 1 in 500	Reduced
9	ANKS6	Nephronophthisis 16	< 1 in 500	Reduced
3	ANO10	Spinocerebellar ataxia, autosomal recessive, type 10	1 in 224	1 in 2236
11	ANO5	Limb-girdle muscular dystrophy, type 12 (LGMD R12)	< 1 in 500	Reduced
2	ANTXR1	GAPO syndrome	< 1 in 500	Reduced
4	ANTXR2	Hyaline fibromatosis syndrome	< 1 in 500	Reduced
7	AP1S1	MEDNIK syndrome	< 1 in 500	Reduced
5	AP3B1	Hermansky-Pudlak syndrome, type 2	< 1 in 500	Reduced
15	AP3B2	Epileptic encephalopathy, early infantile, type 48	< 1 in 500	Reduced
19	AP3D1	Hermansky-Pudlak syndrome, type 10	< 1 in 500	Reduced
1	AP4B1	Spastic paraplegia, type 47, autosomal recessive	< 1 in 500	Reduced
15	AP4E1	Spastic paraplegia, type 51, autosomal recessive	< 1 in 500	Reduced
7	AP4M1	Spastic paraplegia, type 50, autosomal recessive	< 1 in 500	Reduced
14	AP4S1	Spastic paraplegia, type 52, autosomal recessive	1 in 500	1 in 50000
7	AP5Z1	Spastic paraplegia, type 48, autosomal recessive	< 1 in 500	Reduced
19	APOC2	Hyperlipoproteinemia, type 1B	< 1 in 500	Reduced
19	APOE	Sea-blue histiocyte disease	< 1 in 500	Reduced
16	APRT	Adenine phosphoribosyltransferase deficiency	1 in 250	1 in 1667
9	APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	< 1 in 500	Reduced
12	AQP2	Diabetes insipidus, nephrogenic, type 2	< 1 in 500	Reduced
20	ARFGEF2	Periventricular heterotopia with microcephaly	< 1 in 500	Reduced
6	ARG1	Argininemia (arginase deficiency)	1 in 418	1 in 13933
17	ARHGDIA	Nephrotic syndrome, type 8	< 1 in 500	Reduced
19	ARHGEF18	Retinitis pigmentosa 78	< 1 in 500	Reduced

3	ARL13B	Joubert syndrome type 8	< 1 in 500	Reduced
16	ARL2BP	Retinitis pigmentosa with or without situs inversus	< 1 in 500	Reduced
3	ARL6	Bardet-Biedl syndrome, type 3	< 1 in 500	Reduced
2	ARMC9	Joubert syndrome 30	< 1 in 500	Reduced
7	ARPC1B	Immunodeficiency, type 71, with inflammatory disease and congenital thrombocytopenia	< 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
1	ARV1	Epileptic encephalopathy, early infantile, 38	< 1 in 500	Reduced
8	ASAH1	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy	< 1 in 500	Reduced
7	ASL	Argininosuccinic aciduria	1 in 116	1 in 1170
7	ASNS	Asparagine synthetase deficiency	< 1 in 500	Reduced
17	ASPA	Canavan disease	1 in 416	1 in 13867
8	ASPH	Traboulsi syndrome	< 1 in 500	Reduced
1	ASPM	Primary microcephaly type 5, autosomal recessive	< 1 in 500	Reduced
9	ASS1	Citrullinemia, type 1	1 in 300	1 in 3750
10	ATAD1	Hyperekplexia 4	< 1 in 500	Reduced
1	ATF6	Achromatopsia, type 7	< 1 in 500	Reduced
2	ATIC	AICA-ribosiduria due to ATIC deficiency	< 1 in 500	Reduced
11	ATM	ATM-related conditions	1 in 150	1 in 1000
10	ATOH7	Persistent hyperplastic primary vitreous, autosomal recessive	< 1 in 500	Reduced
1	ATP13A2	Kufor-Rakeb syndrome; Spastic paraplegia, type 78, autosomal recessive	1 in 221	1 in 7360
16	ATP2A1	Brody myopathy	< 1 in 500	Reduced
12	ATP6V0A2	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome	< 1 in 500	Reduced
7	ATP6V0A4	Renal tubular acidosis, distal, autosomal recessive	< 1 in 500	Reduced
3	ATP6V1A	Cutis laxa, autosomal recessive, type 2D	< 1 in 500	Reduced
2	ATP6V1B1	Renal tubular acidosis with deafness	< 1 in 500	Reduced
22	ATP6V1E1	Cutis laxa, autosomal recessive, type 2C	< 1 in 500	Reduced
13	ATP7B	Wilson disease	1 in 90	1 in 450
18	ATP8B1	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1	1 in 738	1 in 1803
3	ATR	Seckel syndrome, type 1	< 1 in 500	Reduced
9	AUH	3-methylglutaconic aciduria, type 1	< 1 in 500	Reduced
19	AURKC	Spermatogenic failure, type 5	< 1 in 500	Reduced
12	AVIL	Nephrotic syndrome, type 21	< 1 in 500	Reduced
15	B2M	Immunodeficiency, type 43	< 1 in 500	Reduced
1	B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	< 1 in 500	Reduced
1	B3GALT6	Ehlers-Danlos syndrome, spondylodysplastic type, 2	< 1 in 500	Reduced
11	B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	< 1 in 500	Reduced
13	B3GLCT	Peters-plus syndrome	< 1 in 500	Reduced
12	B4GALNT1	Spastic paraplegia, type 26, autosomal recessive	< 1 in 500	Reduced
9	B4GALT1	Congenital disorder of glycosylation, type 2D	< 1 in 500	<1 in 50000
5	B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic, type 1	< 1 in 500	Reduced
11	B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13	< 1 in 500	Reduced
17	B9D1	Joubert syndrome, type 27; ?Meckel syndrome 9	< 1 in 500	Reduced
19	B9D2	Joubert syndrome, type 34; ?Meckel syndrome, type 10	< 1 in 500	Reduced
11	BBS1	Bardet-Biedl syndrome, type 1	1 in 152	1 in 1520
12	BBS10	Bardet-Biedl syndrome, type 10	1 in 237	1 in 4740
4	BBS12	Bardet-Biedl syndrome, type 12	1 in 500	1 in 10000
16	BBS2	Bardet-Biedl syndrome, type 2	1 in 200	1 in 4000
15	BBS4	Bardet-Biedl syndrome, type 4	1 in 418	1 in 8360
2	BBS5	Bardet-Biedl syndrome, type 5	1 in 500	Reduced
4	BBS7	Bardet-Biedl syndrome, type 7	1 in 500	1 in 5000
7	BBS9	Bardet-Biedl syndrome, type 9	1 in 500	1 in 2500
19	BCAT2	?Hypervalinemia or hyperleucine-isoleucinemia	< 1 in 500	Reduced
3	BCHE	Butyrylcholinesterase deficiency	1 in 30	1 in 200
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
16	BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency	< 1 in 500	Reduced
1	BCL10	?Immunodeficiency, type 37	< 1 in 500	Reduced
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
20	BFSP1	Cataract 33, multiple types	< 1 in 500	Reduced
17	BHLHA9	Syndactyly, mesoaxial synostotic, with phalangeal reduction	< 1 in 500	Reduced
2	BIN1	Centronuclear myopathy, type 2	< 1 in 500	Reduced
15	BLM	Bloom syndrome	1 in 320	1 in 3200
10	BLNK	?Agammaglobulinemia 4	< 1 in 500	Reduced
19	BLOC1S3	Hermansky-Pudlak syndrome, type 8	< 1 in 500	Reduced
15	BLOC1S6	?Hermansky-Pudlak syndrome, type 9	< 1 in 500	Reduced
4	BLTP1	Alkuraya-Kucinskas syndrome	< 1 in 500	Reduced
7	BLVRA	Hyperbiliverdinemia	< 1 in 500	Reduced

8	BMP1	Osteogenesis imperfecta, type 13	1 in 600	1 in 20000
7	BMPER	Diaphanospondylodysostosis	< 1 in 500	Reduced
4	BMPR1B	Acromesomelic dysplasia, Demirhan type	< 1 in 500	Reduced
2	BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia	< 1 in 500	Reduced
7	BPGM	Erythrocytosis due to bisphosphoglycerate mutase deficiency	< 1 in 500	Reduced
8	BPNT2	Chondrodysplasia with joint dislocations, GPAPP type	< 1 in 500	Reduced
7	BRAT1	Rigidity and multifocal seizure syndrome, lethal neonatal; Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	< 1 in 500	Reduced
14	BRF1	Cerebellofaciodental syndrome	< 1 in 500	Reduced
17	BRIP1	Fanconi anemia, complementation group J	< 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 6000
15	BUB1B	Mosaic variegated aneuploidy syndrome 1	< 1 in 500	Reduced
12	C12ORF57	Temtamy syndrome	< 1 in 500	Reduced
19	C19ORF12	Neurodegeneration with brain iron accumulation, type 4	< 1 in 500	Reduced
1	C1QA	C1q deficiency	< 1 in 500	Reduced
1	C1QB	C1q deficiency	< 1 in 500	Reduced
17	C1QBP	Combined oxidative phosphorylation deficiency 33	< 1 in 500	Reduced
1	C1QC	C1q deficiency 3	< 1 in 500	Reduced
12	C1S	C1s deficiency	< 1 in 500	Reduced
6	C2	C2 deficiency	< 1 in 500	1 in 16666
11	C2CD3	Orofaciodigital syndrome, type 14	< 1 in 500	1 in 10000
19	C3	Complement component 3 deficiency	< 1 in 500	Reduced
9	C5	Complement component 5 deficiency	< 1 in 500	Reduced
5	C6	Complement component 6 deficiency	< 1 in 500	Reduced
5	C7	Complement component 7 deficiency	< 1 in 500	Reduced
1	C8B	Complement component 8 deficiency, type 2	< 1 in 500	Reduced
15	CA12	Hyperchlorhidrosis, isolated	< 1 in 500	Reduced
8	CA2	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	< 1 in 500	<1 in 1000
16	CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency	< 1 in 500	Reduced
8	CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	< 1 in 500	Reduced
11	CABP2	Deafness, autosomal recessive, type 93	< 1 in 500	Reduced
11	CABP4	Congenital stationary night blindness, type 2B	< 1 in 500	Reduced
3	CACNA1D	Sinoatrial node dysfunction and deafness	< 1 in 500	Reduced
12	CACNA2D4	Retinal cone dystrophy 4	< 1 in 500	Reduced
2	CAD	Epileptic encephalopathy, early infantile, 50	< 1 in 500	Reduced
2	CALCRL	?Lymphatic malformation 8	< 1 in 500	Reduced
17	CANT1	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7	< 1 in 500	1 in 10000
11	CAPN1	Spastic paraplegia, type 76, autosomal recessive	< 1 in 500	Reduced
15	CAPN3	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	1 in 103	1 in 412
7	CARD11	Immunodeficiency, type 11A	< 1 in 500	Reduced
9	CARD9	Candidiasis, familial, type 2, autosomal recessive	< 1 in 500	Reduced
13	CARS2	Combined oxidative phosphorylation deficiency 27	< 1 in 500	Reduced
1	CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, type 2	1 in 168	1 in 8354
3	CASR	Hyperparathyroidism, neonatal	< 1 in 500	Reduced
5	CAST	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads	< 1 in 500	Reduced
11	CAT	Acatalasemia	< 1 in 500	Reduced
11	CATSPER1	Spermatogenic failure, type 7	< 1 in 500	Reduced
17	CAVIN1	Lipodystrophy, congenital generalized, type 4	< 1 in 500	Reduced
11	CBLIF	Intrinsic factor deficiency	< 1 in 500	Reduced
21	CBS	Homocystinuria due to cystathionine beta-synthase	1 in 274	1 in 2740
19	CC2D1A	Mental retardation, autosomal recessive, type 3	< 1 in 500	Reduced
4	CC2D2A	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2	1 in 196	1 in 2800
18	CCBE1	Hennekam lymphangiectasia-lymphedema syndrome, type 1	< 1 in 500	Reduced
17	CCDC103	Ciliary dyskinesia, primary, type 17	< 1 in 500	Reduced
2	CCDC115	Congenital disorder of glycosylation, type IIo	< 1 in 500	Reduced
3	CCDC174	Hypotonia, infantile, with psychomotor retardation	< 1 in 500	Reduced
3	CCDC39	Ciliary dyskinesia, primary, type 14	1 in 250	1 in 2500
17	CCDC40	Ciliary dyskinesia, primary, type 15	< 1 in 500	1 in 10000
12	CCDC65	Ciliary dyskinesia, primary, type 27	< 1 in 500	Reduced
19	CCDC8	3M syndrome 3	< 1 in 500	Reduced
14	CCDC88C	Hydrocephalus, congenital, type 1	1 in 500	1 in 7143
6	CCN6	Progressive pseudorheumatoid dysplasia	< 1 in 500	Reduced
5	CCNO	Ciliary dyskinesia, primary, type 29	< 1 in 500	Reduced
16	CD19	Immunodeficiency, common variable, type 3	< 1 in 500	Reduced
1	CD247	?Immunodeficiency, type 25	< 1 in 500	Reduced
12	CD27	Lymphoproliferative syndrome 2	< 1 in 500	Reduced
6	CD2AP	Glomerulosclerosis, focal segmental, type 3, susceptibility to	< 1 in 500	Reduced
19	CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect	< 1 in 500	Reduced

7	CD36	Platelet glycoprotein 4 deficiency	< 1 in 500	Reduced
11	CD3D	Immunodeficiency, type 19	< 1 in 500	Reduced
11	CD3E	Immunodeficiency, type 18	< 1 in 500	Reduced
11	CD3G	Immunodeficiency, type 17, CD3 gamma deficient	< 1 in 500	Reduced
20	CD40	Immunodeficiency with hyper-IgM, type 3	< 1 in 500	Reduced
1	CD55	Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy (CHAPLE)	< 1 in 500	Reduced
11	CD59	CD59 deficiency	< 1 in 500	Reduced
19	CD79A	Agammaglobulinemia 3	< 1 in 500	Reduced
17	CD79B	Agammaglobulinemia 6	< 1 in 500	Reduced
11	CD81	Immunodeficiency, common variable, type 6	< 1 in 500	Reduced
2	CD8A	CD8 deficiency, familial	< 1 in 500	Reduced
15	CDAN1	Dyserythropoietic anemia, congenital, type 1A	1 in 500	1 in 50000
1	CDC14A	Deafness, autosomal recessive, type 105	< 1 in 500	Reduced
22	CDC45	Meier-Gorlin syndrome 7	< 1 in 500	Reduced
2	CDCA7	Immunodeficiency-centromeric instability-facial anomalies syndrome 3	< 1 in 500	Reduced
16	CDH11	Elsahy-Waters syndrome	< 1 in 500	Reduced
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	< 1 in 500	Reduced
10	CDHR1	Cone-rod dystrophy, type 15	< 1 in 500	Reduced
15	CDIN1	Dyserythropoietic anemia, congenital, type Ib	< 1 in 500	Reduced
16	CDK10	Al Kaissi syndrome	< 1 in 500	Reduced
9	CDK5RAP2	Primary microcephaly type 3, autosomal recessive	< 1 in 500	Reduced
6	CDSN	Peeling skin syndrome 1	< 1 in 500	Reduced
16	CDT1	Meier-Gorlin syndrome, type 4	< 1 in 500	Reduced
14	CEBPE	Specific granule deficiency	< 1 in 500	Reduced
1	CENPF	Stromme syndrome	< 1 in 500	Reduced
13	CENPJ	Primary microcephaly type 6, autosomal recessive	< 1 in 500	Reduced
1	CEP104	Joubert syndrome 25	< 1 in 500	Reduced
5	CEP120	Short-rib thoracic dysplasia 13 with or without polydactyly	< 1 in 500	Reduced
4	CEP135	Microcephaly 8, primary, autosomal recessive	1 in 500	1 in 5000
15	CEP152	Primary microcephaly type 9, autosomal recessive	< 1 in 500	Reduced
11	CEP164	Nephronophthisis 15	< 1 in 500	Reduced
3	CEP19	Morbid obesity and spermatogenic failure	< 1 in 500	Reduced
12	CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	1 in 150	1 in 375
7	CEP41	Joubert syndrome, type 15	< 1 in 500	Reduced
10	CEP55	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly	< 1 in 500	Reduced
11	CEP57	Mosaic variegated aneuploidy syndrome 2	< 1 in 500	Reduced
9	CEP78	Cone-rod dystrophy and hearing loss	< 1 in 500	Reduced
12	CEP83	Nephronophthisis 18	< 1 in 500	Reduced
2	CERKL	Retinitis pigmentosa, type 26	1 in 250	1 in 1667
15	CERS3	Ichthyosis, congenital, autosomal recessive 9	< 1 in 500	Reduced
8	CFAP418	Bardet-Biedl syndrome, type 21; Cone-rod dystrophy 16 and Retintis pigmentosa 64; Ciliary dyskinesia, primary, 26	< 1 in 500	Reduced
10	CFAP43	Spermatogenic failure, type 19	< 1 in 500	Reduced
18	CFAP53	Heterotaxy, visceral, 6, autosomal recessive	< 1 in 500	Reduced
19	CFD	Complement factor D deficiency	< 1 in 500	Reduced
1	CFH	Complement factor H deficiency	< 1 in 500	Reduced
4	CFI	Complement factor I deficiency	1 in 500	1 in 50000
14	CFL2	Nemaline myopathy, type 7, autosomal recessive	< 1 in 500	Reduced
7	CFTR	Cystic fibrosis	1 in 25	1 in 833
10	CHAT	Myasthenic syndrome, congenital, type 6, presynaptic	1 in 600	1 in 30000
22	CHKB	Muscular dystrophy, congenital, megaconial type	< 1 in 500	Reduced
16	CHMP1A	Pontocerebellar hypoplasia, type 8	< 1 in 500	Reduced
2	CHRNA1	Multiple pterygium syndrome, lethal type	1 in 500	1 in 50000
17	CHRNB1	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	< 1 in 500	Reduced
2	CHRND	Myasthenic syndrome, congenital, type 3B, fast-channel; Multiple pterygium syndrome, lethal type	< 1 in 500	Reduced
17	CHRNE	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	1 in 244	1 in 2440
2	CHRNG	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	1 in 400	1 in 1597
15	CHST14	Ehlers-Danlos syndrome, musculocontractural, type 1	< 1 in 500	Reduced
10	CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations	< 1 in 500	Reduced
16	CHST6	Macular corneal dystrophy	1 in 80	1 in 394
15	CHSY1	Temtamy preaxial brachydactyly syndrome	< 1 in 500	Reduced
10	CHUK	Cocoon syndrome	< 1 in 500	Reduced
15	CIB2	Deafness, autosomal recessive, type 48; Usher syndrome, type 1J	< 1 in 500	Reduced
16	CIITA	Bare lymphocyte syndrome, type 2, complementation group A	< 1 in 500	Reduced
6	CILK1	Endocrine-cerebroosteodysplasia	< 1 in 500	Reduced
4	CISD2	Wolfram syndrome 2	< 1 in 500	Reduced
12	CIT	Microcephaly 17, primary, autosomal recessive	< 1 in 500	Reduced
2	CKAP2L	Filippi syndrome	< 1 in 500	Reduced

11	CLCF1	Cold-induced sweating syndrome 2	< 1 in 500	Reduced
7	CLCN1	Myotonia congenita, recessive	1 in 159	1 in 319
3	CLCN2	Leukoencephalopathy with ataxia	< 1 in 500	Reduced
16	CLCN7	Osteopetrosis, autosomal recessive type 4	< 1 in 500	Reduced
1	CLCNKA	Bartter syndrome, type 4B, digenic	< 1 in 500	Reduced
1	CLCNKB	Bartter syndrome, type 3; Bartter syndrome, type 4B, digenic	< 1 in 500	1 in 1000
3	CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	< 1 in 500	Reduced
13	CLDN10	HELIX syndrome	< 1 in 500	Reduced
21	CLDN14	Deafness type 29, autosomal recessive	< 1 in 500	Reduced
3	CLDN16	Hypomagnesemia, type 3, renal	< 1 in 500	Reduced
1	CLDN19	Rena hypomagnesemia type 5, with ocular involvement	< 1 in 500	Reduced
11	CLMP	Congenital short bowel syndrome	< 1 in 500	Reduced
16	CLN3	Ceroid lipofuscinosis, neuronal, type 3	1 in 242	1 in 2090
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
11	CLP1	Pontocerebellar hypoplasia, type 10	< 1 in 500	Reduced
11	CLPB	3-methylglutaconic aciduria, type 7, with cataracts, neurologic involvement and neutropenia	< 1 in 500	Reduced
19	CLPP	Perrault syndrome 3	< 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 1171
2	CNGA3	Achromatopsia, type 2	1 in 500	1 in 50000
16	CNGB1	Retinitis pigmentosa type 45	1 in 200	1 in 4000
8	CNGB3	Achromatopsia, type 3	1 in 125	1 in 1250
10	CNNM2	Hypomagnesemia, seizures, and mental retardation	< 1 in 500	Reduced
2	CNNM4	Jalili syndrome	< 1 in 500	Reduced
6	CNPY3	Epileptic encephalopathy, early infantile, type 60	< 1 in 500	Reduced
17	CNTNAP1	Lethal congenital contracture syndrome 7	< 1 in 500	Reduced
7	CNTNAP2	Pitt-Hopkins like syndrome 1	< 1 in 500	Reduced
1	COA6	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4	< 1 in 500	Reduced
14	COA8	Mitochondrial complex IV deficiency, nuclear type 17	< 1 in 500	Reduced
17	COASY	Neurodegeneration with brain iron accumulation 6	< 1 in 500	Reduced
17	COG1	Congenital disorder of glycosylation, type IIg	< 1 in 500	Reduced
16	COG4	Congenital disorder of glycosylation, type 2J	< 1 in 500	Reduced
7	COG5	Congenital disorder of glycosylation, type 2I	< 1 in 500	Reduced
13	COG6	Congenital disorder of glycosylation, type 2L; Shaheen syndrome	< 1 in 500	Reduced
16	COG7	Congenital disorder of glycosylation, type 2E	< 1 in 500	Reduced
16	COG8	Congenital disorder of glycosylation, type 2H	< 1 in 500	Reduced
1	COL11A1	Fibrochondrogenesis type 1	1 in 500	1 in 16666
6	COL11A2	Otospondylomegaepiphyseal dysplasia, autosomal recessive	1 in 500	1 in 16666
10	COL13A1	Myasthenic syndrome, congenital, 19	< 1 in 500	Reduced
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	< 1 in 500	Reduced
4	COL25A1	Fibrosis of extraocular muscles, congenital, type 5	< 1 in 500	Reduced
9	COL27A1	Steel syndrome	1 in 500	1 in 2500
2	COL4A3	Alport syndrome, autosomal recessive, type 3B	1 in 300	1 in 1500
2	COL4A4	Alport syndrome, autosomal recessive, type 2	1 in 425	1 in 4250
21	COL6A1	Ullrich congenital muscular dystrophy, type 1 (Limb-girdle muscular dystrophy, type 22 [LGMD R22])	< 1 in 500	1 in 25000
21	COL6A2	Ullrich congenital muscular dystrophy 1B; Bethlem myopathy-1B; Myosclerosis	< 1 in 500	1 in 25000
2	COL6A3	Bethlem myopathy 1; Ullrich congenital muscular dystrophy 1; Dystonia 27	< 1 in 500	1 in 25000
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	< 1 in 500	Reduced
1	COL9A2	?Stickler syndrome, type V	< 1 in 500	Reduced
8	COLEC10	3MC syndrome 3	< 1 in 500	Reduced
2	COLEC11	3MC syndrome 2	< 1 in 500	Reduced
3	COLQ	Myasthenic syndrome, congenital, type 5	1 in 805	1 in 1420
4	COQ2	Primary coenzyme Q10 deficiency, type 1	< 1 in 500	Reduced
9	COQ4	Coenzyme Q10 deficiency, primary, type 7	< 1 in 500	Reduced
14	COQ6	Coenzyme Q10 deficiency, primary, type 6	< 1 in 500	Reduced
1	COQ8A	Primary coenzyme Q10 deficiency, type 4	< 1 in 500	Reduced
19	COQ8B	Nephrotic syndrome, type 9	< 1 in 500	Reduced
16	COQ9	Coenzyme Q10 deficiency, primary, type 5	< 1 in 500	Reduced
16	CORO1A	Immunodeficiency, type 8	< 1 in 500	Reduced
17	COX10	Mitochondrial complex IV deficiency, nuclear type 3	< 1 in 500	Reduced
10	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 9600
1	COX20	Mitochondrial complex IV deficiency, nuclear type 11	< 1 in 500	Reduced
19	COX6B1	Mitochondrial complex IV deficiency, nuclear type 7	< 1 in 500	Reduced
3	CP	Aceruloplasminemia	< 1 in 500	Reduced

8	CPA6	Febrile seizures, familial, type 11	< 1 in 500	Reduced
19	CPAMD8	Anterior segment dysgenesis, type 8	< 1 in 500	Reduced
5	CPLANE1	Joubert syndrome 17	1 in 150	1 in 7500
4	CPLX1	Epileptic encephalopathy, early infantile, 63	< 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	CR2	Immunodeficiency, common variable, type 7	< 1 in 500	Reduced
12	CRADD	Mental retardation, autosomal recessive, type 34, with variant lissencephaly	< 1 in 500	Reduced
1	CRB1	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	1 in 158	1 in 1580
9	CRB2	Ventriculomegaly with cystic kidney disease	1 in 500	1 in 16600
3	CRBN	Mental retardation, autosomal recessive, type 2	< 1 in 500	Reduced
2	CRIPT	Short stature with microcephaly and distinctive facies	< 1 in 500	Reduced
19	CRLF1	Cold-induced sweating syndrome type 1	< 1 in 500	Reduced
7	CRPPA	Muscular dystrophy-dystroglycanopathy, type A7; Muscular dystrophy-dystroglycanopathy, type C7	< 1 in 500	Reduced
3	CRTAP	Osteogenesis imperfecta, type 7	1 in 1416	1 in 3539
21	CRYAA	Cataract 9, multiple types	< 1 in 500	Reduced
11	CRYAB	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related; Cataract 16, multiple types	< 1 in 500	Reduced
22	CRYBB1	Cataract 17	< 1 in 500	Reduced
22	CRYBB3	Cataract 22	< 1 in 500	Reduced
22	CSF2RB	Surfactant metabolism dysfunction, pulmonary, type 5	< 1 in 500	Reduced
1	CSF3R	Neutropenia, severe congenital, type 7, autosomal recessive	< 1 in 500	Reduced
8	CSPP1	Joubert syndrome 21	< 1 in 500	Reduced
3	CSTA	Peeling skin syndrome, type 4	< 1 in 500	Reduced
21	CSTB	Epilepsy, progressive myoclonic type 1A (Unverricht and Lundborg)	< 1 in 500	Reduced
17	CTC1	Cerebroretinal microangiopathy with calcifications and cysts	< 1 in 500	Reduced
1	CTH	Cystathioninuria	1 in 160	1 in 3200
17	CTNS	Nephropathic cystinosis	1 in 200	1 in 400
1	CTPS1	Immunodeficiency, type 24	< 1 in 500	Reduced
20	CTSA	Galactosialidosis	1 in 557	1 in 11140
11	CTSC	Haim-Munk syndrome; Papillon-Lefevre syndrome	1 in 500	1 in 2496
11	CTSD	Ceroid lipofuscinosis, neuronal, type 10	< 1 in 500	Reduced
11	CTSF	Ceroid lipofuscinosis, neuronal, type 13 (Kufs type)	< 1 in 500	Reduced
1	CTSK	Pycnodysostosis	< 1 in 500	Reduced
10	CUBN	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)	1 in 500	1 in 10000
6	CUL7	3M syndrome 1	< 1 in 500	Reduced
5	CWC27	Retinitis pigmentosa with or without skeletal anomalies	< 1 in 500	Reduced
10	CWF19L1	Spinocerebellar ataxia, autosomal recessive, type 17	< 1 in 500	Reduced
18	CYB5A	46,XY disorder of sex development due to isolated 17,20-lyase deficiency	< 1 in 500	Reduced
22	CYB5R3	Methemoglobinemia, type 1; Methemoglobinemia, type 2	< 1 in 500	Reduced
16	CYBA	Chronic granulomatous disease, type 4	< 1 in 500	Reduced
8	CYC1	Mitochondrial complex III deficiency, nuclear type 6	< 1 in 500	Reduced
15	CYP11A1	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	1 in 500	1 in 7143
8	CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	1 in 300	1 in 2000
8	CYP11B2	Hypoaldosteronism, congenital, due to CMO I deficiency	< 1 in 500	Reduced
10	CYP17A1	17 alpha(α)-hydroxylase/17,20-lyase deficiency	< 1 in 500	Reduced
15	CYP19A1	Aromatase deficiency	< 1 in 500	Reduced
2	CYP1B1	Glaucoma, primary congenital, type 3A	1 in 196	1 in 1960
6	CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	1 in 62	1 in 1240
20	CYP24A1	Hypercalcemia, infantile, type 1	1 in 500	1 in 16600
2	CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies	< 1 in 500	Reduced
10	CYP26C1	Focal facial dermal dysplasia 4	< 1 in 500	Reduced
2	CYP27A1	Cerebrotendinous xanthomatosis	1 in 275	1 in 5500
12	CYP27B1	Vitamin D-dependent rickets, type 1	< 1 in 500	Reduced
11	CYP2R1	Rickets due to defect in vitamin D 25-hydroxylation	< 1 in 500	Reduced
4	CYP2U1	Spastic paraplegia, type 56, autosomal recessive	< 1 in 500	Reduced
19	CYP4F22	Ichthyosis, congenital, autosomal recessive, type 5	< 1 in 500	Reduced
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
3	DAG1	Muscular dystrophy-dystroglycanopathy type A9; Muscular dystrophy-dystroglycanopathy type C9	< 1 in 500	Reduced
2	DARS1	Hypomyelination with brainstem and spinal cord involvement and leg spasticity	< 1 in 500	Reduced
1	DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	< 1 in 500	Reduced
9	DBH	Dopamine beta-hydroxylase deficiency	< 1 in 500	Reduced
1	DBT	Maple syrup urine disease, type 2	1 in 410	1 in 2733
2	DCAF17	Woodhouse-Sakati syndrome	< 1 in 500	Reduced
18	DCC	Gaze palsy, familial horizontal, with progressive scoliosis, type 2	< 1 in 500	Reduced
6	DCDC2	Sclerosing cholangitis, neonatal; Nephronophthisis 19	< 1 in 500	Reduced
11	DCHS1	Van Maldergem syndrome 1	< 1 in 500	Reduced

10	DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	< 1 in 500	Reduced
11	DCPS	Al-Raqad syndrome	< 1 in 500	Reduced
11	DDB2	Xeroderma pigmentosum, complementation group E	< 1 in 500	Reduced
7	DDC	Aromatic L-amino acid decarboxylase deficiency	< 1 in 500	Reduced
14	DDHD1	Spastic paraplegia, type 28, autosomal recessive	< 1 in 500	Reduced
8	DDHD2	Spastic paraplegia, type 54, autosomal recessive	< 1 in 500	Reduced
1	DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type	< 1 in 500	Reduced
20	DDRGK1	Spondyloepimetaphyseal dysplasia, Shohat type	< 1 in 500	Reduced
12	DDX11	Warsaw breakage syndrome	< 1 in 500	Reduced
1	DDX59	Orofaciodigital syndrome V	< 1 in 500	Reduced
11	DENND5A	Epileptic encephalopathy, early infantile, 49	< 1 in 500	Reduced
2	DES	Myopathy, myofibrillar, type 1	< 1 in 500	Reduced
8	DGAT1	?Diarrhea 7, protein-losing enteropathy type	1 in 500	1 in 8300
17	DGKE	Nephrotic syndrome, type 7	< 1 in 500	Reduced
2	DGUOK	DGUOK-related mitochondrial DNA depletion syndrome	< 1 in 500	Reduced
1	DHCR24	Desmosterolosis	< 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
5	DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency	< 1 in 500	Reduced
12	DHH	46,XY complete gonadal dysgenesis	< 1 in 500	Reduced
16	DHODH	Miller syndrome	< 1 in 500	Reduced
19	DHPS	Neurodevelopmental disorder with seizures and speech and walking impairment	< 1 in 500	Reduced
10	DHTKD1	2-aminoadipic 2-oxoadipic aciduria	< 1 in 500	Reduced
5	DIAPH1	Seizures, cortical blindness, microcephaly syndrome	< 1 in 500	Reduced
2	DIS3L2	Perlman syndrome	< 1 in 500	Reduced
11	DLAT	Pyruvate dehydrogenase E2 deficiency	< 1 in 500	Reduced
7	DLD	Dihydrolipoamide dehydrogenase deficiency	< 1 in 500	Reduced
19	DLL3	Spondylocostal dysostosis type 1	< 1 in 500	1 in 5000
5	DMGDH	Dimethylglycine dehydrogenase deficiency	< 1 in 500	Reduced
4	DMP1	Hypophosphatemic rickets, autosomal recessive	< 1 in 500	Reduced
15	DMXL2	Developmental and epileptic encephalopathy, type 81	< 1 in 500	Reduced
16	DNAAF1	Ciliary dyskinesia, primary, type 13	< 1 in 500	Reduced
8	DNAAF11	Ciliary dyskinesia, primary, type 19	< 1 in 500	Reduced
14	DNAAF2	Ciliary dyskinesia, primary, type 10	< 1 in 500	Reduced
19	DNAAF3	Ciliary dyskinesia, primary, type 2	< 1 in 500	Reduced
15	DNAAF4	Ciliary dyskinesia, primary, type 25	< 1 in 500	Reduced
7	DNAAF5	Ciliary dyskinesia, primary, type 18	< 1 in 500	Reduced
3	DNAH1	Spermatogenic failure, type 18	< 1 in 500	Reduced
7	DNAH11	Ciliary dyskinesia, primary, type 7, with or without situs inversus	1 in 500	1 in 2500
5	DNAH5	Ciliary dyskinesia, primary, type 3, with or without situs inversus	1 in 130	1 in 520
17	DNAH9	Ciliary dyskinesia, primary, type 40	< 1 in 500	Reduced
9	DNAI1	Ciliary dyskinesia, primary, type 1, with or without situs inversus	1 in 323	1 in 1615
17	DNAI2	Ciliary dyskinesia, primary, type 9, with or without situs inversus	< 1 in 500	Reduced
11	DNAJB13	Ciliary dyskinesia, primary, type 34	< 1 in 500	Reduced
2	DNAJB2	Spinal muscular atrophy, distal, autosomal recessive, type 5	< 1 in 500	Reduced
10	DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient	1 in 500	Reduced
3	DNAJC19	3-methylglutaconic aciduria, type 5	< 1 in 500	Reduced
5	DNAJC21	Bone marrow failure syndrome, type 3	< 1 in 500	Reduced
1	DNAJC6	Parkinson disease, type 19A, juvenile-onset; Parkinson disease, type 19B, early-onset	< 1 in 500	Reduced
14	DNAL1	Ciliary dyskinesia, primary, type 16	< 1 in 500	Reduced
3	DNASE1L3	Systemic lupus erythematosus 16	< 1 in 500	Reduced
12	DNM1L	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 1	< 1 in 500	Reduced
19	DNM2	Lethal congenital contracture syndrome, type 5	< 1 in 500	Reduced
20	DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 1	< 1 in 500	Reduced
5	DOCK2	Immunodeficiency, type 40	< 1 in 500	Reduced
19	DOCK6	Adams-Oliver syndrome 2	< 1 in 500	Reduced
1	DOCK7	Epileptic encephalopathy, early infantile, 23	< 1 in 500	Reduced
9	DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive	< 1 in 500	Reduced
4	DOK7	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	1 in 262	1 in 2620
9	DOLK	Congenital disorder of glycosylation, type 1M	< 1 in 500	Reduced
21	DONSON	Microcephaly, short stature, and limb abnormalities	< 1 in 500	Reduced
11	DPAGT1	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	< 1 in 500	Reduced
17	DPH1	Developmental delay with short stature, dysmorphic features, and sparse hair	< 1 in 500	Reduced
20	DPM1	Congenital disorder of glycosylation, type 1E	< 1 in 500	<1 in 1750
9	DPM2	Congenital disorder of glycosylation, type Iu	< 1 in 500	Reduced
1	DPM3	Congenital disorder of glycosylation, type Io	< 1 in 500	Reduced
12	DPY19L2	Spermatogenic failure, type 9	< 1 in 500	Reduced
1	DPYD	Dihydropyrimidine dehydrogenase deficiency	1 in 558	1 in 55701
8	DPYS	Dihydropyrimidinuria	< 1 in 500	Reduced

1	DRAM2	Cone-rod dystrophy 21	< 1 in 500	Reduced
2	DRC1	Ciliary dyskinesia, primary, type 21	< 1 in 500	Reduced
18	DSG1	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE	< 1 in 500	Reduced
18	DSG4	Hypotrichosis, type 6	< 1 in 500	Reduced
6	DSP	Cardiomyopathy, dilated, with woolly hair and keratoderma; Epidermolysis bullosa, lethal acantholytic	< 1 in 500	Reduced
6	DST	Neuropathy, hereditary sensory and autonomic, type VI;Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency	< 1 in 500	Reduced
1	DSTYK	Spastic paraplegia, type 23, autosomal recessive	< 1 in 500	Reduced
6	DTNBP1	Hermansky-Pudlak syndrome, type 7	< 1 in 500	Reduced
15	DUOX2	Thyroid dyshormonogenesis, type 6	1 in 80	1 in 2600
15	DUOXA2	Thyroid dyshormonogenesis, type 5	1 in 80	1 in 800
18	DYM	Smith-McCort dysplasia; Dyggve-Melchior-Clausen disease	< 1 in 500	Reduced
11	DYNC2H1	Short-rib thoracic dysplasia, type 3, with or without polydactyly	1 in 50	1 in 500
7	DYNC2I1	Short-rib thoracic dysplasia 8 with or without polydactyly	< 1 in 500	Reduced
9	DYNC2I2	Short-rib thoracic dysplasia 11 with or without polydactyly	< 1 in 500	Reduced
2	DYNC2LI1	Short-rib thoracic dysplasia 15 with polydactyly	< 1 in 500	Reduced
3	DYNLT2B	Short-rib thoracic dysplasia 17 with or without polydactyly	< 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
3	DZIP1L	Polycystic kidney disease 5	< 1 in 500	Reduced
16	EARS2	Combined oxidative phosphorylation deficiency 12	< 1 in 500	Reduced
2	ECEL1	Arthrogryposis, distal, type 5D	< 1 in 500	Reduced
10	ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	< 1 in 500	Reduced
1	ECM1	Urbach-Wiethe disease	1 in 500	1 in 50000
2	EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	1 in 746	1 in 14920
1	EDARADD	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type	< 1 in 500	Reduced
6	EDN1	Auriculocondylar syndrome, type 3	< 1 in 500	Reduced
20	EDN3	Waardenburg syndrome, type 4B	< 1 in 500	Reduced
13	EDNRB	ABCD syndrome	< 1 in 500	Reduced
11	EFEMP2	Cutis laxa, autosomal recessive, type 1B	< 1 in 500	Reduced
15	EFL1	Shwachman-Diamond syndrome 2	< 1 in 500	Reduced
7	EGFR	?Inflammatory skin and bowel disease, neonatal, 2	< 1 in 500	Reduced
10	EGR2	Dejerine-Sottas disease	< 1 in 500	Reduced
2	EIF2AK3	Wolcott-Rallison syndrome	< 1 in 500	<1 in 2500
15	EIF2AK4	Pulmonary venoocclusive disease 2	< 1 in 500	Reduced
12	EIF2B1	Leukoencephalopathy with vanishing white matter (VWM)	< 1 in 500	Reduced
14	EIF2B2	Leukoencephalopathy with vanishing white matter (VWM)	< 1 in 500	Reduced
1	EIF2B3	Leukoencephalopathy with vanishing white matter (VWM)	< 1 in 500	Reduced
2	EIF2B4	Leukoencephalopathy with vanishing white matter (VWM)	< 1 in 500	Reduced
3	EIF2B5	Leukoencephalopathy with vanishing white matter (VWM)	1 in 400	1 in 8000
17	EIF4A3	Robin sequence with cleft mandible and limb anomalies	< 1 in 500	Reduced
17	ELAC2	Combined oxidative phosphorylation deficiency 17	< 1 in 500	Reduced
20	ELMO2	Vascular malformation, primary intraosseous	< 1 in 500	Reduced
6	ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation	< 1 in 500	Reduced
9	ELP1	Familial dysautonomia	1 in 200	1 in 2000
18	ELP2	Mental retardation, autosomal recessive, type 58	< 1 in 500	Reduced
1	EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation	< 1 in 500	Reduced
14	EML1	Band heterotopia	< 1 in 500	Reduced
16	EMP2	Nephrotic syndrome, type 10	< 1 in 500	Reduced
4	ENAM	Amelogenesis imperfecta, type 1C	< 1 in 500	Reduced
17	ENO3	?Glycogen storage disease XIII	< 1 in 500	1 in 10000
6	ENPP1	Arterial calcification, generalized, of infancy, type 1	1 in 333	1 in 3330
10	ENTPD1	Spastic paraplegia, type 64, autosomal recessive	< 1 in 500	Reduced
3	EOGT	Adams-Oliver syndrome 4	< 1 in 500	Reduced
1	EPB41	Elliptocytosis, type 1	< 1 in 500	Reduced
15	EPB42	Spherocytosis, type 5	< 1 in 500	Reduced
2	EPCAM	EPCAM-related conditions	< 1 in 500	Reduced
18	EPG5	Vici syndrome	< 1 in 500	Reduced
6	EPM2A	Epilepsy, progressive myoclonic, type 2A (Lafora)	1 in 250	1 in 1600
1	EPRS1	Leukodystrophy, hypomyelinating, type 15	< 1 in 500	Reduced
11	EPS8L2	Deafness autosomal recessive, type 106	< 1 in 500	Reduced
17	ERAL1	Perrault syndrome 6	< 1 in 500	Reduced
12	ERBB3	Lethal congenital contractural syndrome, type 2	< 1 in 500	Reduced
19	ERCC1	Cerebrooculofacioskeletal syndrome, type 4	< 1 in 500	Reduced
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 1306
16	ERCC4	Fanconi anemia, complementation group Q	< 1 in 500	Reduced
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
9	ERCC6L2	Bone marrow failure syndrome, type 2	< 1 in 500	Reduced
5	ERCC8	Cockayne syndrome, type A	1 in 514	1 in 3960

10	ERLIN1	Spastic paraplegia, type 62, autosomal recessive	< 1 in 500	Reduced
8	ERLIN2	Spastic paraplegia, type 18, autosomal recessive	< 1 in 500	Reduced
8	ESCO2	Roberts syndrome	< 1 in 500	Reduced
1	ESPN	Deafness, autosomal recessive, type 36	< 1 in 500	Reduced
6	ESR1	Estrogen resistance	< 1 in 500	Reduced
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
4	EVC	Ellis-van Creveld syndrome	1 in 240	1 in 960
4	EVC2	Ellis-van Creveld syndrome	1 in 300	1 in 2000
9	EXOSC3	Pontocerebellar hypoplasia, type 1B	1 in 139	1 in 300
11	EXPH5	Epidermolysis bullosa, nonspecific, autosomal recessive	< 1 in 500	Reduced
8	EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities	< 1 in 500	Reduced
6	EYS	Retinitis pigmentosa, type 25	1 in 100	1 in 1000
13	F10	Factor X deficiency	< 1 in 500	Reduced
4	F11	Factor XI deficiency	1 in 200	1 in 2500
6	F13A1	Factor XIIIA deficiency	< 1 in 500	Reduced
1	F13B	Factor XIIIB deficiency	< 1 in 500	Reduced
11	F2	Prothrombin deficiency	1 in 300	1 in 1500
1	F5	Factor V deficiency	1 in 500	1 in 10000
13	F7	Factor VII deficiency	1 in 500	1 in 7100
16	FA2H	Spastic paraplegia, type 35, autosomal recessive	< 1 in 500	Reduced
11	FADD	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations	< 1 in 500	Reduced
15	FAH	Tyrosinemia, type 1	1 in 200	1 in 2000
2	FAM161A	Retinitis pigmentosa, type 28	1 in 350	1 in 3500
17	FAM20A	Amelogenesis imperfecta, type 1G (Enamel-renal syndrome)	< 1 in 500	Reduced
7	FAM20C	Raine syndrome	< 1 in 500	<1 in 1000
15	FAN1	Interstitial nephritis, karyomegalic	< 1 in 500	Reduced
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
3	FANCD2	Fanconi anemia, complementation group D2	< 1 in 500	Reduced
6	FANCE	Fanconi anemia, complementation group E	< 1 in 500	Reduced
11	FANCF	Fanconi anemia, complementation group F	< 1 in 500	Reduced
9	FANCG	Fanconi anemia, complementation group G	< 1 in 500	Reduced
15	FANCI	Fanconi anemia, complementation group I	< 1 in 500	Reduced
2	FANCL	Fanconi anemia, complementation group L	1 in 400	1 in 4000
14	FANCM	Spermatogenic failure, type 28; ?Premature ovarian failure 15	< 1 in 500	Reduced
11	FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder	< 1 in 500	Reduced
6	FARS2	Combined oxidative phosphorylation deficiency 14; Spastic paraplegia, type 77, autosomal recessive	< 1 in 500	Reduced
2	FASTKD2	Combined oxidative phosphorylation deficiency 44	< 1 in 500	Reduced
4	FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2	< 1 in 500	Reduced
14	FBLN5	Cutis laxa, autosomal recessive, type 1A	< 1 in 500	Reduced
9	FBP1	Fructose-1,6-bisphosphatase deficiency	< 1 in 500	Reduced
6	FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	< 1 in 500	Reduced
22	FBXO7	Parkinson disease, type 15, autosomal recessive	< 1 in 500	Reduced
17	FDXR	Auditory neuropathy and optic atrophy	< 1 in 500	Reduced
18	FECH	Protoporphyria, erythropoietic, autosomal recessive	1 in 500	1 in 3333
20	FERMT1	Kindler syndrome	< 1 in 500	Reduced
11	FERMT3	Leukocyte adhesion deficiency, type 3	< 1 in 500	Reduced
7	FEZF1	Hypogonadotropic hypogonadism type 22, with or without anosmia	< 1 in 500	Reduced
4	FGA	Afibrinogenemia, congenital	< 1 in 500	Reduced
4	FGB	Congenital afibrinogenemia	< 1 in 500	Reduced
12	FGD4	Charcot-Marie-Tooth disease, type 4H	< 1 in 500	Reduced
12	FGF23	Tumoral calcinosis, hyperphosphatemic, familial, type 2	< 1 in 500	Reduced
11	FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia	< 1 in 500	Reduced
4	FGG	Afibrinogenemia, congenital; Hypofibrinogenemia, congenital	< 1 in 500	Reduced
1	FH	Fumarase deficiency	1 in 500	1 in 3333
11	FIBP	Thauvin-Robinet-Faivre syndrome	< 1 in 500	Reduced
6	FIG4	Charcot-Marie-Tooth disease, type 4J; Yunis-Varon syndrome	< 1 in 500	Reduced
17	FKBP10	Bruck syndrome 1	< 1 in 500	Reduced
7	FKBP14	Ehlers-Danlos syndrome, kyphoscoliotic type, 2	< 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	FLAD1	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	< 1 in 500	Reduced
1	FLG	Ichthyosis vulgaris	1 in 200	1 in 10000
11	FLI1	Bleeding disorder, platelet-type, type 21	< 1 in 500	Reduced
3	FLNB	Spondylocarpotarsal synostosis syndrome	< 1 in 500	Reduced

1	FLVCR1	Posterior column ataxia-retinitis pigmentosa syndrome	< 1 in 500	Reduced
14	FLVCR2	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	< 1 in 500	Reduced
1	FMN2	Mental retardation, autosomal recessive, type 47	< 1 in 500	Reduced
1	FMO3	Trimethylaminuria	1 in 100	1 in 1000
11	FOLR1	Neurodegeneration due to cerebral folate transport deficiency	< 1 in 500	Reduced
9	FOXE1	Bamforth-Lazarus syndrome	< 1 in 500	Reduced
1	FOXE3	Anterior segment dysgenesis, type 2, multiple subtypes	< 1 in 500	Reduced
17	FOXN1	T-cell immunodeficiency, congenital alopecia and nail dystrophy	< 1 in 500	Reduced
11	FOXRED1	Mitochondrial complex I deficiency, nuclear type 19	1 in 439	1 in 21950
4	FRAS1	Fraser syndrome, type 1	1 in 300	1 in 3000
9	FREM1	Manitoba oculotrichoanal syndrome	< 1 in 500	Reduced
13	FREM2	Fraser syndrome, type 2	1 in 115	1 in 3833
9	FRRS1L	Epileptic encephalopathy, early infantile, 37	< 1 in 500	Reduced
11	FSHB	Hypogonadotropic hypogonadism, type 24, without anosmia	< 1 in 500	Reduced
2	FSHR	Ovarian dysgenesis 1;Ovarian hyperstimulation syndrome;Ovarian response to FSH stimulation	< 1 in 500	Reduced
21	FTCD	Glutamate formiminotransferase deficiency	1 in 355	1 in 7000
19	FTL	L-ferritin deficiency	< 1 in 500	Reduced
16	FTO	Growth retardation, developmental delay, facial dysmorphism	< 1 in 500	Reduced
1	FUCA1	Fucosidosis	1 in 1149	1 in 4880
14	FUT8	Congenital disorder of glycosylation with defective fucosylation, type 1	< 1 in 500	Reduced
9	FXN	Friedreich ataxia	1 in 91	1 in 1014
3	FYCO1	Cataract 18	< 1 in 500	1 in 25000
8	FZD6	Nail disorder, nonsyndromic congenital, type 10 (claw-shaped nails)	< 1 in 500	Reduced
17	G6PC1	Glycogen storage disease, type 1A	1 in 300	1 in 3000
17	G6PC3	Dursun syndrome	< 1 in 500	<1 in 1170
17	GAA	Glycogen storage disease, type 2	1 in 100	1 in 500
14	GALC	Krabbe disease	1 in 120	1 in 218
1	GALE	Galactose epimerase deficiency	< 1 in 500	Reduced
17	GALK1	Galactokinase deficiency with cataracts	1 in 200	1 in 2500
16	GALNS	Mucopolysaccharidosis, type 4A	1 in 311	1 in 5180
2	GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, type 1	1 in 500	1 in 10000
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
16	GAS8	Ciliary dyskinesia, primary, type 33	< 1 in 500	Reduced
15	GATM	Cerebral creatine deficiency syndrome, type 3	< 1 in 500	Reduced
1	GBA1	Gaucher Disease, type I-III; GD IIIC; GD, perinatal lethal	1 in 125	1 in 1563
9	GBA2	Spastic paraplegia, type 46, autosomal recessive	< 1 in 500	Reduced
3	GBE1	Glycogen storage disease, type 4	1 in 192	1 in 960
19	GCDH	Glutaricaciduria, type 1	1 in 200	1 in 4000
14	GCH1	Hyperphenylalaninemia, BH4-deficient, type B	1 in 250	1 in 1600
7	GCK	Permanent neonatal diabetes mellitus (PNDM)	< 1 in 500	Reduced
6	GCM2	Hypoparathyroidism, familial isolated (FIH) 2	< 1 in 500	Reduced
6	GCNT2	Cataract 13, with adult i phenotype	< 1 in 500	Reduced
16	GCSH	Multiple mitochondrial dysfunctions syndrome 7	< 1 in 500	Reduced
8	GDAP1	Charcot-Marie-Tooth disease, recessive intermediate, type A	1 in 180	1 in 9000
19	GDF1	Right atrial isomerism (Ivemark syndrome)	1 in 500	1 in 16600
20	GDF5	Chondrodysplasia, Grebe type	< 1 in 500	Reduced
8	GDF6	Leber congenital amaurosis, type 17	< 1 in 500	Reduced
16	GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	< 1 in 500	Reduced
3	GFM1	Combined oxidative phosphorylation deficiency, type 1	1 in 450	1 in 1500
2	GFPT1	Myasthenia, congenital, type 12, with tubular aggregates	< 1 in 500	Reduced
2	GGCX	Vitamin K-dependent clotting factors, combined deficiency of, type 1	< 1 in 500	Reduced
17	GH1	Growth hormone deficiency, isolated, type 1A; Kowarski syndrome	< 1 in 500	Reduced
5	GHR	Laron dwarfism	< 1 in 500	Reduced
7	GHRHR	Growth hormone deficiency, isolated, type 1B	1 in 692	1 in 34560
3	GHSR	Growth hormone deficiency, isolated partial	< 1 in 500	Reduced
20	GINS1	Immunodeficiency, type 55	< 1 in 500	Reduced
19	GIPC3	Deafness, autosomal recessive, type 15	< 1 in 500	Reduced
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 4210
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
15	GLDN	Lethal congenital contracture syndrome 11	< 1 in 500	Reduced
9	GLE1	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	1 in 350	1 in 3500
16	GLIS2	Nephronophthisis, type 7	< 1 in 500	Reduced
9	GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism	< 1 in 500	Reduced

5	GLRA1	Hyperekplexia, type 1	< 1 in 500	Reduced
4	GLRB	Hyperekplexia, type 2	< 1 in 500	Reduced
14	GLRX5	Anemia, sideroblastic, type 3, pyridoxine-refractory; Spasticity, childhood-onset, with hyperglycinemia	< 1 in 500	Reduced
1	GLUL	Glutamine deficiency, congenital	< 1 in 500	Reduced
3	GLYCTK	D-glyceric aciduria	< 1 in 500	Reduced
5	GM2A	GM2-gangliosidosis, AB variant	< 1 in 500	Reduced
2	GMPPA	Alacrima, achalasia, and mental retardation syndrome	< 1 in 500	Reduced
3	GMPPB	Muscular dystrophy-dystroglycanopathy 14	1 in 500	1 in 25000
3	GNAT1	Night blindness, congenital stationary, type 1G	< 1 in 500	Reduced
1	GNAT2	Achromatopsia, type 4	< 1 in 500	Reduced
15	GNB5	Intellectual developmental disorder with cardiac arrhythmia; Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia	< 1 in 500	Reduced
9	GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)	1 in 203	1 in 4060
6	GNMT	Glycine N-methyltransferase deficiency	1 in 500	1 in 24951
1	GNPAT	Rhizomelic chondrodysplasia punctata, type 2	< 1 in 500	Reduced
12	GNPTAB	Mucolipidosis 2 alpha/beta; Mucolipidosis 3 alpha/beta	1 in 176	1 in 17522
16	GNPTG	Mucolipidosis III gamma	< 1 in 500	Reduced
4	GNRHR	Hypogonadotropic hypogonadism, type 7, without anosmia	1 in 500	1 in 2097
12	GNS	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	< 1 in 500	Reduced
1	GORAB	Geroderma osteodysplasticum	< 1 in 500	Reduced
17	GOSR2	Epilepsy, progressive myoclonic, type 6	< 1 in 500	Reduced
16	GOT2	Epileptic encephalopathy, early infantile, 82	< 1 in 500	Reduced
17	GP1BA	Bernard-Soulier syndrome, type A1	< 1 in 500	Reduced
22	GP1BB	Bernard-Soulier syndrome, type B	1 in 500	1 in 531
19	GP6	Bleeding disorder, platelet-type, type 11	< 1 in 500	Reduced
3	GP9	Bernard-Soulier syndrome, type C	1 in 451	1 in 4510
8	GPAA1	Glycosylphosphatidylinositol biosynthesis defect 15	< 1 in 500	Reduced
13	GPC6	Omodysplasia, type 1	< 1 in 500	Reduced
12	GPD1	Hypertriglyceridemia, transient infantile	< 1 in 500	Reduced
14	GPHN	Molybdenum cofactor deficiency C	< 1 in 500	Reduced
19	GPI	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency	< 1 in 500	Reduced
8	GPIHBP1	Hyperlipoproteinemia, type 1D	< 1 in 500	Reduced
17	GPR179	Night blindness, congenital stationary (complete), type 1E, autosomal recessive	< 1 in 500	Reduced
14	GPR68	Amelogenesis imperfecta, type 2A6 (hypomaturation type)	< 1 in 500	Reduced
1	GPSM2	Chudley-McCullough syndrome	< 1 in 500	Reduced
16	GPT2	Mental retardation, autosomal recessive 49	< 1 in 500	Reduced
19	GPX4	Spondylometaphyseal dysplasia, Sedaghatian type	< 1 in 500	Reduced
8	GRHL2	Ectodermal dysplasia/short stature syndrome	< 1 in 500	Reduced
9	GRHPR	Hyperoxaluria, primary, type 2	1 in 433	1 in 21650
4	GRID2	Spinocerebellar ataxia, autosomal recessive, type 18	< 1 in 500	Reduced
6	GRIK2	Mental retardation, autosomal recessive, type, 6	< 1 in 500	Reduced
9	GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive	< 1 in 500	Reduced
12	GRIP1	Fraser syndrome 3	1 in 1218	1 in 12180
13	GRK1	Oguchi disease-2	< 1 in 500	Reduced
6	GRM1	Spinocerebellar ataxia, autosomal recessive, type 13	< 1 in 500	Reduced
5	GRM6	Night blindness, congenital stationary (complete), type 1B, autosomal recessive	< 1 in 500	Reduced
17	GRN	Ceroid lipofuscinosis, neuronal, type 11	< 1 in 500	Reduced
4	GRXCR1	Deafness, autosomal recessive, type 25	< 1 in 500	Reduced
14	GSC	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities	< 1 in 500	Reduced
20	GSS	Glutathione synthetase deficiency	< 1 in 500	Reduced
6	GTF2H5	Trichothiodystrophy, type 3, photosensitive	< 1 in 500	Reduced
6	GTPBP2	Jaberi-Elahi syndrome	< 1 in 500	Reduced
19	GTPBP3	Combined oxidative phosphorylation deficiency 23	< 1 in 500	Reduced
12	GUCY2C	Meconium ileus	< 1 in 500	Reduced
17	GUCY2D	Leber congenital amaurosis, type 1	1 in 248	1 in 305
4	GUF1	?Epileptic encephalopathy, early infantile, 40	< 1 in 500	1 in 10000
7	GUSB	Mucopolysaccharidosis, type 7	1 in 552	1 in 16531
3	GYG1	Polyglucosan body myopathy, type 2	< 1 in 500	1 in 10000
19	GYS1	Glycogen storage disease, type 0, muscle	< 1 in 500	Reduced
12	GYS2	Glycogen storage disease, type 0, liver	< 1 in 500	Reduced
20	GZF1	Joint laxity, short stature, and myopia	< 1 in 500	Reduced
1	H6PD	Cortisone reductase deficiency 1	< 1 in 500	Reduced
2	HAAO	Vertebral, cardiac, renal, and limb defects syndrome 1	< 1 in 500	Reduced
6	HACE1	Spastic paraplegia and psychomotor retardation with or without seizures	< 1 in 500	Reduced
4	HADH	3-hydroxyacyl-CoA dehydrogenase deficiency	1 in 415	1 in 622
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
19	HAMP	Hemochromatosis, type 2B	< 1 in 500	Reduced
5	HARS1	Usher syndrome, type 3B	< 1 in 500	Reduced
1	HAX1	Neutropenia, severe congenital, type 3, autosomal recessive	1 in 219	1 in 2190

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 6700
10	HELLS	Immunodeficiency-centromeric instability-facial anomalies syndrome 4	< 1 in 500	Reduced
11	HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A	< 1 in 500	Reduced
15	HERC1	Macrocephaly, dysmorphic facies, and psychomotor retardation	< 1 in 500	Reduced
15	HERC2	Mental retardation, autosomal recessive, type 38	< 1 in 500	1 in 5000
17	HES7	Spondylocostal dysostosis, type 4, autosomal recessive	< 1 in 500	Reduced
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
1	HFM1	Premature ovarian failure 9	< 1 in 500	Reduced
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	< 1 in 500	Reduced
11	HIKESHI	Leukodystrophy, hypomyelinating, type 13	< 1 in 500	Reduced
5	HINT1	Neuromyotonia and axonal neuropathy, autosomal recessive	< 1 in 500	Reduced
1	HJV	Hemochromatosis, type 2A	< 1 in 500	Reduced
10	HK1	Charcot-Marie-Tooth disease, type 4G	< 1 in 500	Reduced
21	HLCS	Holocarboxylase synthetase deficiency	1 in 300	1 in 3000
1	HMGCL	HMG-CoA lyase deficiency	< 1 in 500	Reduced
1	HMGCS2	HMG-CoA synthase-2 deficiency	< 1 in 500	Reduced
22	HMOX1	Heme oxygenase-1 deficiency	1 in 500	Reduced
4	HMX1	Oculoauricular syndrome	< 1 in 500	Reduced
2	HNMT	Mental retardation, autosomal recessive, type 51	< 1 in 500	Reduced
10	HOGA1	Hyperoxaluria, primary, type 3	1 in 169	1 in 8400
7	HOXA1	Athabaskan brainstem dysgenesis syndrome	< 1 in 500	Reduced
17	HOXB1	Facial paresis, hereditary congenital, 3	< 1 in 500	Reduced
12	HOXC13	Ectodermal dysplasia 9, hair/nail type	< 1 in 500	Reduced
1	HPCA	Dystonia 2, torsion, autosomal recessive	< 1 in 500	Reduced
12	HPD	Tyrosinemia, type 3	< 1 in 500	1 in 16666
4	HPGD	Hypertrophic osteoarthropathy, primary, type 1 (pachydermoperiostosis)	< 1 in 500	Reduced
10	HPS1	Hermansky-Pudlak syndrome, type 1	1 in 493	1 in 4930
3	HPS3	Hermansky-Pudlak syndrome, type 3	1 in 300	1 in 1500
22	HPS4	Hermansky-Pudlak syndrome, type 4	< 1 in 500	1 in 16666
11	HPS5	Hermansky-Pudlak syndrome, type 5	< 1 in 500	1 in 16666
10	HPS6	Hermansky-Pudlak syndrome, type 6	1 in 400	1 in 13333
10	HPSE2	Urofacial syndrome, type 1	< 1 in 500	Reduced
8	HR	Alopecia universalis; Atrichia with papular lesions	< 1 in 500	Reduced
16	HSD11B2	Apparent mineralocorticoid excess	< 1 in 500	Reduced
9	HSD17B3	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	< 1 in 500	<1 in 2750
5	HSD17B4	D-bifunctional protein deficiency	1 in 534	1 in 13350
1	HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	1 in 862	1 in 8620
16	HSD3B7	Bile acid synthesis defect, congenital, type 1	< 1 in 500	Reduced
5	HSPA9	Even-plus syndrome	< 1 in 500	Reduced
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 1625
10	HTRA1	CARASIL syndrome	< 1 in 500	Reduced
2	HTRA2	3-methylglutaconic aciduria, type 8	< 1 in 500	Reduced
3	HYAL1	?Mucopolysaccharidosis, type 9	< 1 in 500	Reduced
7	HYCC1	Leukodystrophy, hypomyelinating, type 5	< 1 in 500	Reduced
16	HYDIN	Ciliary dyskinesia, primary, type 5	< 1 in 500	1 in 2380
11	HYLS1	Hydrolethalus syndrome	1 in 500	1 in 714
9	IARS1	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy	< 1 in 500	Reduced
1	IBA57	Multiple mitochondrial dysfunctions syndrome 3	< 1 in 500	Reduced
2	ICOS	Immunodeficiency, common variable, 1	< 1 in 500	Reduced
20	IDH3B	Retinitis pigmentosa, type 46	1 in 500	1 in 999
4	IDUA	Mucopolysaccharidosis type 1	1 in 153	1 in 3825
18	IER3IP1	Microcephaly, epilepsy, and diabetes syndrome	< 1 in 500	Reduced
6	IFNGR1	Immunodeficiency, type 27A, mycobacteriosis	< 1 in 500	Reduced
21	IFNGR2	Immunodeficiency, type 28, mycobacteriosis	< 1 in 500	Reduced
3	IFT122	Cranioectodermal dysplasia 1	< 1 in 500	Reduced
16	IFT140	Retinitis pigmentosa, type 80; Short-rib thoracic dysplasia 9 with or without polydactyly	1 in 500	1 in 952
2	IFT172	Short-rib thoracic dysplasia 10 with or without polydactyly	< 1 in 500	Reduced
14	IFT43	Short-rib thoracic dysplasia 18 with polydactyly	< 1 in 500	Reduced
20	IFT52	Short-rib thoracic dysplasia 16 with or without polydactyly	< 1 in 500	Reduced
3	IFT80	Short-rib thoracic dysplasia, type 2, with or without polydactyly	< 1 in 500	Reduced
12	IFT81	Short-rib thoracic dysplasia 19 with or without polydactyly	< 1 in 500	Reduced

12	IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency	< 1 in 500	Reduced
15	IGF1R	Insulin-like growth factor I, resistance to	< 1 in 500	Reduced
16	IGFALS	Acid-labile subunit deficiency	< 1 in 500	Reduced
4	IGFBP7	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	< 1 in 500	Reduced
11	IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S	< 1 in 500	<1 in 4000
22	IGLL1	Agammaglobulinemia 2	< 1 in 500	Reduced
2	IHH	Acrocapitofemoral dysplasia	< 1 in 500	Reduced
8	IKBKB	Immunodeficiency, type 15	< 1 in 500	Reduced
11	IL10RA	Inflammatory bowel disease, type 28, early onset, autosomal recessive	< 1 in 500	Reduced
21	IL10RB	Inflammatory bowel disease, type 25, early onset, autosomal recessive	< 1 in 500	Reduced
9	IL11RA	Craniosynostosis and dental anomalies	< 1 in 500	Reduced
5	IL12B	Immunodeficiency, type 29, mycobacteriosis	< 1 in 500	Reduced
19	IL12RB1	Immunodeficiency, type 30	< 1 in 500	Reduced
22	IL17RA	Immunodeficiency, type 51	< 1 in 500	Reduced
3	IL17RC	Candidiasis, familial, 9	< 1 in 500	Reduced
2	IL1RN	Sterile multifocal osteomyelitis with periostitis and pustulosis	< 1 in 500	Reduced
16	IL21R	Immunodeficiency, type 56	< 1 in 500	Reduced
10	IL2RA	Immunodeficiency, type 41, with lymphoproliferation and autoimmunity	< 1 in 500	Reduced
2	IL36RN	Psoriasis, type 14, pustular	< 1 in 500	Reduced
5	IL7R	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type	< 1 in 500	1 in 5000
3	ILDR1	Deafness, autosomal recessive, type 42	< 1 in 500	Reduced
8	IMPA1	Mental retardation, autosomal recessive 59	< 1 in 500	Reduced
3	IMPG2	Retinitis pigmentosa, type 56	< 1 in 500	Reduced
9	INPP5E	Joubert syndrome, type 1	< 1 in 500	Reduced
17	INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability	< 1 in 500	Reduced
11	INPPL1	Opsismodysplasia	< 1 in 500	Reduced
11	INS	Permanent neonatal diabetes mellitus (PNDM)	< 1 in 500	Reduced
19	INSR	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, type A	< 1 in 500	Reduced
7	INTS1	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies	< 1 in 500	Reduced
9	INVS	Nephronophthisis, type 2, infantile	1 in 500	1 in 10000
3	IQCB1	Senior-Loken syndrome, type 5	< 1 in 500	Reduced
7	IQCE	Polydactyly, postaxial, type A7	1 in 500	1 in 5000
12	IRAK4	Immunodeficiency, type 67 (IRAK4 deficiency)	< 1 in 500	Reduced
16	IRF8	Immunodeficiency, type 32B, monocyte and dendritic cell deficiency	< 1 in 500	Reduced
16	IRX5	Hamamy syndrome	< 1 in 500	Reduced
9	ISCA1	Multiple mitochondrial dysfunctions syndrome 5	< 1 in 500	Reduced
14	ISCA2	Multiple mitochondrial dysfunctions syndrome 4	< 1 in 500	Reduced
12	ISCU	Myopathy with lactic acidosis, hereditary	< 1 in 500	Reduced
1	ISG15	Immunodeficiency, type 38	< 1 in 500	Reduced
20	ITCH	Autoimmune disease, multisystem, with facial dysmorphism	< 1 in 500	Reduced
17	ITGA2B	Glanzmann thrombasthenia	1 in 500	1 in 10000
17	ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	1 in 500	1 in 10000
2	ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis	< 1 in 500	Reduced
12	ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency	< 1 in 500	Reduced
10	ITGA8	Renal hypodysplasia/aplasia 1	< 1 in 500	Reduced
21	ITGB2	Leukocyte adhesion deficiency	< 1 in 500	Reduced
17	ITGB3	Glanzmann thrombasthenia	1 in 500	1 in 12500
17	ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia	< 1 in 500	Reduced
2	ITGB6	Amelogenesis imperfecta, type 1H	1 in 500	1 in 16600
5	ITK	Lymphoproliferative syndrome 1	< 1 in 500	Reduced
20	ITPA	Epileptic encephalopathy, early infantile, type 35	< 1 in 500	Reduced
3	ITPR1	Gillespie syndrome	< 1 in 500	Reduced
15	IVD	Isovaleric acidemia	1 in 115	1 in 1917
6	IYD	Thyroid dyshormonogenesis, type 4	1 in 159	1 in 633
3	JAGN1	Neutropenia, severe congenital, 6, autosomal recessive	< 1 in 500	Reduced
19	JAK3	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	1 in 475	1 in 2375
11	JAM3	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	< 1 in 500	Reduced
17	JUP	Naxos disease	< 1 in 500	Reduced
19	KANK2	Nephrotic syndrome, type 16	< 1 in 500	Reduced
16	KARS1	Deafness, autosomal recessive, type 89	< 1 in 500	Reduced
16	KATNB1	Lissencephaly 6, with microcephaly	< 1 in 500	Reduced
16	KATNIP	Joubert syndrome 26	< 1 in 500	Reduced
21	KCNE1	Jervell and Lange-Nielsen syndrome 2	< 1 in 500	Reduced
11	KCNJ1	Bartter syndrome, type 2	< 1 in 500	Reduced
1	KCNJ10	SESAME syndrome	< 1 in 500	Reduced
11	KCNJ11	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	1 in 232	1 in 4640
2	KCNJ13	Leber congenital amaurosis, type 16	< 1 in 500	Reduced
9	KCNV2	Retinal cone dystrophy, type 3B	< 1 in 500	Reduced
7	KCTD7	Epilepsy, progressive myoclonic, type 3, with or without intracellular inclusions	< 1 in 500	Reduced

18	KDSR	Erythrokeratodermia variabilis et progressiva 4	< 1 in 500	Reduced
12	KERA	Cornea plana 2, autosomal recessive	< 1 in 500	Reduced
6	KHDC3L	Hydatidiform mole, recurrent, type 2	< 1 in 500	Reduced
14	KIAA0586	Joubert syndrome 23; Short-rib thoracic dysplasia 14 with polydactyly	1 in 500	1 in 2500
17	KIAA0753	?Orofaciodigital syndrome, type 15	< 1 in 500	Reduced
7	KIAA1549	Retinitis pigmentosa, type 86	< 1 in 500	Reduced
1	KIF14	Microcephaly 20, primary, autosomal recessive; ?Meckel syndrome 12	< 1 in 500	Reduced
2	KIF1A	Neuropathy, hereditary sensory, type 2C; Spastic paraplegia, type 30, autosomal recessive	< 1 in 500	Reduced
17	KIF1C	Spastic ataxia 2, autosomal recessive	< 1 in 500	Reduced
15	KIF7	Acrocallosal syndrome; Joubert syndrome, type 12	< 1 in 500	Reduced
10	KIFBP	Goldberg-Shprintzen megacolon syndrome	< 1 in 500	Reduced
19	KISS1R	Hypogonadotropic hypogonadism, type 8, with or without anosmia	< 1 in 500	Reduced
20	KIZ	Retinitis pigmentosa 69	< 1 in 500	Reduced
5	KLHL3	Pseudohypoaldosteronism, type 2D	< 1 in 500	Reduced
3	KLHL40	Nemaline myopathy 8, autosomal recessive	< 1 in 500	Reduced
2	KLHL41	Nemaline myopathy 9	< 1 in 500	Reduced
7	KLHL7	Cold-induced sweating syndrome 3	< 1 in 500	Reduced
19	KLK4	Amelogenesis imperfecta, type 2A1 (hypomaturation type)	< 1 in 500	Reduced
4	KLKB1	Fletcher factor (prekallikrein) deficiency	< 1 in 500	Reduced
15	KNL1	Microcephaly 4, primary, autosomal recessive	< 1 in 500	Reduced
19	KPTN	Mental retardation, autosomal recessive 41	< 1 in 500	Reduced
22	KREMEN1	Ectodermal dysplasia 13, hair/tooth type	< 1 in 500	Reduced
17	KRT10	Epidermolytic hyperkeratosis 2B, autosomal recessive	< 1 in 500	Reduced
17	KRT14	Epidermolysis bullosa simplex, autosomal recessive, type 1	< 1 in 500	Reduced
17	KRT25	Woolly hair, autosomal recessive 3	< 1 in 500	Reduced
12	KRT5	Epidermolysis bullosa simplex, autosomal recessive, type 1	< 1 in 500	Reduced
12	KRT85	Ectodermal dysplasia 4, hair/nail type	< 1 in 500	Reduced
3	KY	Myopathy, myofibrillar, type 7	< 1 in 500	Reduced
2	KYNU	Vertebral, cardiac, renal, and limb defects syndrome, type 2	< 1 in 500	Reduced
14	L2HGDH	L-2-hydroxyglutaric aciduria	< 1 in 500	Reduced
18	LAMA1	Poretti-Boltshauser syndrome	< 1 in 500	<1 in 1178
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
7	LAMB1	Lissencephaly, type 5	< 1 in 500	Reduced
3	LAMB2	Pierson syndrome; Nephrotic syndrome, type 5, with or without ocular abnormalities	< 1 in 500	1 in 16666
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
9	LAMC3	Cortical malformations, occipital	< 1 in 500	Reduced
22	LARGE1	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	1 in 123	1 in 410
4	LARP7	Alazami syndrome	< 1 in 500	Reduced
5	LARS1	?Infantile liver failure syndrome 1 (ILFS1)	< 1 in 500	Reduced
3	LARS2	Perrault syndrome, type 4	< 1 in 500	Reduced
16	LAT	Immunodeficiency, type 52	< 1 in 500	Reduced
1	LBR	Greenberg skeletal dysplasia	< 1 in 500	Reduced
6	LCA5	Leber congenital amaurosis, type 5	< 1 in 500	Reduced
16	LCAT	Familial LCAT deficiency; Fish-eye disease	< 1 in 500	1 in 4166
1	LCK	?Immunodeficiency, type 22	< 1 in 500	Reduced
2	LCT	Lactase deficiency, congenital	< 1 in 500	Reduced
11	LDHA	Glycogen storage disease type 11	< 1 in 500	Reduced
19	LDLR	Hypercholesterolemia, familial, type 1	1 in 118	1 in 280
1	LDLRAP1	Hypercholesterolemia, familial, autosomal recessive	< 1 in 500	Reduced
6	LEMD2	Cataract 46, juvenile-onset	< 1 in 500	Reduced
7	LEP	Obesity, morbid, due to leptin deficiency	< 1 in 500	Reduced
1	LEPR	Obesity, morbid, due to leptin receptor deficiency	< 1 in 500	Reduced
19	LGI4	Arthrogryposis multiplex congenita, neurogenic, with myelin defect	< 1 in 500	Reduced
19	LHB	Hypogonadotropic hypogonadism, type 23, with or without anosmia	< 1 in 500	Reduced
2	LHCGR	Leydig cell hypoplasia	< 1 in 500	<1 in 1060
6	LHFPL5	Deafness, autosomal recessive, type 67	< 1 in 500	Reduced
9	LHX3	Pituitary hormone deficiency, combined, type 3	1 in 1398	1 in 13980
4	LIAS	Hyperglycinemia, lactic acidosis, and seizures	< 1 in 500	Reduced
5	LIFR	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	< 1 in 500	Reduced
13	LIG4	LIG4 syndrome	1 in 580	1 in 11600
19	LIM2	Cataract 19, multiple types	< 1 in 500	Reduced
15	LINS1	Mental retardation, autosomal recessive, type 27	< 1 in 500	Reduced
10	LIPA	Lysosomal acid lipase deficiency	1 in 112	1 in 2240
19	LIPE	Lipodystrophy, familial partial, type 6	< 1 in 500	Reduced
3	LIPH	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	1 in 500	1 in 2995
10	LIPN	Ichthyosis, congenital, autosomal recessive 8	< 1 in 500	Reduced
2	LIPT1	Lipoyltransferase 1 deficiency	< 1 in 500	Reduced

11	LIPT2	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities	< 1 in 500	Reduced
18	LMAN1	Combined deficiency of factor V and factor VIII, type 1	< 1 in 500	Reduced
6	LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	1 in 414	1 in 552
16	LMF1	Lipase deficiency, combined	< 1 in 500	Reduced
3	LMOD3	Nemaline myopathy 10	< 1 in 500	Reduced
19	LONP1	CODAS syndrome	< 1 in 500	Reduced
18	LOXHD1	Deafness, autosomal recessive, type 77	1 in 150	1 in 1500
13	LPAR6	Hypotrichosis, type 8 or woolly hair, autosomal recessive, type 1, with or without hypotrichosis	< 1 in 500	Reduced
2	LPIN1	Myoglobinuria, acute recurrent, autosomal recessive	< 1 in 500	Reduced
18	LPIN2	Majeed syndrome	< 1 in 500	Reduced
8	LPL	Lipoprotein lipase deficiency	1 in 374	1 in 3740
4	LRAT	Leber congenital amaurosis type 14	< 1 in 500	Reduced
4	LRBA	Immunodeficiency, common variable, 8, with autoimmunity	< 1 in 500	Reduced
1	LRIG2	Urofacial syndrome 2	< 1 in 500	Reduced
4	LRIT3	Night blindness, congenital stationary (complete), 1F, autosomal recessive	< 1 in 500	Reduced
10	LRMDA	Albinism, oculocutaneous, type 7	< 1 in 500	Reduced
2	LRP2	Donnai-Barrow syndrome	< 1 in 500	Reduced
11	LRP4	Cenani-Lenz syndactyly syndrome	< 1 in 500	Reduced
11	LRP5	Osteoporosis-pseudoglioma syndrome	< 1 in 500	Reduced
4	LRPAP1	Myopia, type 23, autosomal recessive	< 1 in 500	Reduced
2	LRPPRC	Leigh syndrome, French-Canadian type	< 1 in 500	Reduced
9	LRSAM1	Charcot-Marie-Tooth disease, axonal, type 2P	< 1 in 500	Reduced
11	LRTOMT	Deafness, autosomal recessive, type 63	< 1 in 500	Reduced
21	LSS	Alopecia-intellectual disability syndrome 4; Cataract 44; Hypotrichosis 14	< 1 in 500	Reduced
14	LTBP2	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma	< 1 in 500	Reduced
11	LTBP3	Dental anomalies and short stature	< 1 in 500	Reduced
19	LTBP4	Cutis laxa, autosomal recessive, type 1C	< 1 in 500	Reduced
5	LYRM7	Mitochondrial complex III deficiency, nuclear type 8	< 1 in 500	Reduced
1	LYST	Chediak-Higashi syndrome	< 1 in 500	Reduced
3	LZTFL1	Bardet-Biedl syndrome, type 17	< 1 in 500	Reduced
22	LZTR1	Noonan syndrome, type 2	< 1 in 500	Reduced
19	MAG	Spastic paraplegia, type 75, autosomal recessive	< 1 in 500	Reduced
7	MAGI2	Nephrotic syndrome, type 15	< 1 in 500	Reduced
6	MAK	Retinitis pigmentosa type 62	1 in 1010	1 in 20200
18	MALT1	Immunodeficiency, type 12	< 1 in 500	Reduced
9	MAN1B1	Mental retardation, autosomal recessive, type 15	< 1 in 500	Reduced
19	MAN2B1	Alpha-mannosidosis	1 in 274	1 in 5480
4	MANBA	Mannosidosis, beta	< 1 in 500	Reduced
2	MAP3K20	Centronuclear myopathy, type 6, with fiber-type disproportion	< 1 in 500	Reduced
15	MAPKBP1	Nephronophthisis 20	< 1 in 500	Reduced
17	MAPT	Supranuclear palsy, progressive atypical (parkinsonism syndrome)	< 1 in 500	Reduced
12	MARS1	Interstitial lung and liver disease	< 1 in 500	Reduced
2	MARS2	Spastic ataxia, type 3, autosomal recessive	< 1 in 500	Reduced
5	MARVELD2	Deafness, autosomal recessive, type 49	< 1 in 500	Reduced
3	MASP1	3MC syndrome 1	< 1 in 500	Reduced
10	MAT1A	Methionine adenosyltransferase deficiency, autosomal recessive	< 1 in 500	Reduced
2	MATN3	?Spondyloepimetaphyseal dysplasia	< 1 in 500	Reduced
19	MBOAT7	Mental retardation, autosomal recessive 57	< 1 in 500	Reduced
18	MC2R	Glucocorticoid deficiency, due to ACTH unresponsiveness	< 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 50000
2	MCFD2	Combined deficiency of factor V and factor VIII, type 2	< 1 in 500	Reduced
5	MCIDAS	Ciliary dyskinesia, primary, type 42	< 1 in 500	Reduced
21	MCM3AP	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development	< 1 in 500	Reduced
8	MCM4	Immunodeficiency, type 54	< 1 in 500	Reduced
6	MCM9	Ovarian dysgenesis 4	< 1 in 500	Reduced
19	MCOLN1	Mucolipidosis type 4	1 in 1166	1 in 4850
8	MCPH1	Microcephaly type 1, primary, autosomal recessive	1 in 500	1 in 8333
7	MDH2	Epileptic encephalopathy, early infantile, 51	< 1 in 500	Reduced
1	MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities	< 1 in 500	Reduced
11	MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	< 1 in 500	Reduced
6	MED23	Mental retardation, autosomal recessive, type 18	< 1 in 500	Reduced
19	MED25	Basel-Vanagait-Smirin-Yosef syndrome	< 1 in 500	Reduced
16	MEFV	Familial Mediterranean fever	1 in 40	1 in 133
5	MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	< 1 in 500	Reduced
19	MEGF8	Carpenter syndrome, type 2	< 1 in 500	Reduced
17	MEOX1	Klippel-Feil syndrome 2	< 1 in 500	Reduced
2	MERTK	Retinitis pigmentosa type 38	1 in 500	1 in 2500

15	MESP2	Spondylocostal dysostosis, type 2, autosomal recessive	1 in 500	1 in 50000
17	METTL23	Mental retardation, autosomal recessive 44	< 1 in 500	Reduced
2	MFF	Encephalopathy due to defective mitochondrial and peroxisomal fission, type 2	< 1 in 500	Reduced
1	MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2B	< 1 in 500	1 in 50000
11	MFRP	Microphthalmia, isolated type 5	1 in 250	1 in 1667
1	MFSD2A	Microcephaly 15, primary, autosomal recessive	< 1 in 500	Reduced
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	MGME1	Mitochondrial DNA depletion syndrome 11	< 1 in 500	Reduced
12	MGP	Keutel syndrome	< 1 in 500	Reduced
10	MICU1	Myopathy with extrapyramidal signs	< 1 in 500	Reduced
13	MIPEP	Combined oxidative phosphorylation deficiency 31	< 1 in 500	Reduced
3	MITF	COMMAD syndrome	< 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
2	MLPH	Griscelli syndrome, type 3	< 1 in 500	Reduced
16	MLYCD	Malonyl-CoA decarboxylase deficiency	1 in 500	1 in 1000
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
3	MME	Charcot-Marie-Tooth disease, axonal, type 2T	1 in 500	1 in 16600
11	MMP13	Metaphyseal dysplasia, Spahr type	< 1 in 500	Reduced
16	MMP2	Multicentric osteolysis, nodulosis, and arthropathy (MONA)	< 1 in 500	Reduced
11	MMP20	Amelogenesis imperfecta, type 2A2 (hypomaturation type)	< 1 in 500	Reduced
10	MMP21	Heterotaxy, visceral, 7, autosomal	< 1 in 500	Reduced
6	MMUT	Methylmalonic aciduria, mut(0) type	1 in 135	1 in 3375
18	MOCOS	Xanthinuria, type 2	< 1 in 500	Reduced
6	MOCS1	Molybdenum cofactor deficiency A	1 in 350	1 in 3500
5	MOCS2	Molybdenum cofactor deficiency B	1 in 400	1 in 4000
2	MOGS	Congenital disorder of glycosylation, type 2B	< 1 in 500	Reduced
6	MPC1	Mitochondrial pyruvate carrier deficiency	< 1 in 500	Reduced
17	MPDU1	Congenital disorder of glycosylation, type 1F	< 1 in 500	Reduced
9	MPDZ	Hydrocephalus, congenital, type 2, with or without brain or eye anomalies	< 1 in 500	<1 in 1000
15	MPI	Congenital disorder of glycosylation, type 1B	1 in 473	1 in 11825
6	MPIG6B	Thrombocytopenia, anemia, and myelofibrosis	< 1 in 500	Reduced
1	MPL	Thrombocytopenia, congenital amegakaryocytic	1 in 241	1 in 2410
7	MPLKIP	Trichothiodystrophy, type 4, nonphotosensitive	< 1 in 500	Reduced
17	MPO	Myeloperoxidase deficiency	< 1 in 500	Reduced
2	MPV17	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	1 in 612	1 in 7650
1	MPZ	Dejerine-Sottas disease	< 1 in 500	Reduced
21	MRAP	Glucocorticoid deficiency, type 2	< 1 in 500	Reduced
11	MRE11	Ataxia-telangiectasia-like disorder 1	< 1 in 500	1 in 5000
10	MRPS16	Combined oxidative phosphorylation deficiency 2	< 1 in 500	Reduced
3	MRPS22	Combined oxidative phosphorylation deficiency type 5	< 1 in 500	Reduced
16	MRPS34	Combined oxidative phosphorylation deficiency 32	< 1 in 500	Reduced
5	MSH3	Familial adenomatous polyposis, type 4	< 1 in 500	1 in 10000
4	MSMO1	Microcephaly, congenital cataract, and psoriasiform dermatitis	< 1 in 500	Reduced
12	MSRB3	Deafness, autosomal recessive, type 74	< 1 in 500	Reduced
1	MSTO1	Myopathy, mitochondrial, and ataxia	< 1 in 500	Reduced
15	MTFMT	Combined oxidative phosphorylation deficiency 15	< 1 in 500	1 in 3333
14	MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	< 1 in 500	Reduced
1	MTHFR	Homocystinuria due to MTHFR deficiency	1 in 1119	1 in 11190
11	MTMR2	Charcot-Marie-Tooth disease, type 4B1	1 in 500	1 in 1000
6	MTO1	Combined oxidative phosphorylation deficiency 10	< 1 in 500	Reduced
1	MTR	Homocystinuria-megaloblastic anemia, cblG complementation type	< 1 in 500	1 in 5000
12	MTRFR	Combined oxidative phosphorylation deficiency 7; Spastic paraplegia, type 55, autosomal recessive	< 1 in 500	Reduced
5	MTRR	Homocystinuria-megaloblastic anemia, cbl E type	< 1 in 500	Reduced
4	MTTP	Abetalipoproteinemia	< 1 in 500	Reduced
9	MUSK	Fetal akinesia deformation sequence, type 1; Myasthenic syndrome, congenital, type 9, associated with acetylcholine receptor deficiency	< 1 in 500	Reduced
1	MUTYH	Adenomas, multiple colorectal	1 in 500	1 in 25000
12	MVK	Mevalonic aciduria	1 in 286	1 in 2261
12	MYBPC1	Lethal congenital contracture syndrome, type 4	< 1 in 500	Reduced
3	MYD88	Immunodeficiency, type 68	< 1 in 500	Reduced
17	MYH2	Proximal myopathy and ophthalmoplegia	< 1 in 500	Reduced
9	MYMK	Carey-Fineman-Ziter syndrome	< 1 in 500	Reduced
17	MYO15A	Deafness, autosomal recessive, type 3	1 in 1000	1 in 10000
22	MYO18B	Klippel-Feil syndrome, type 4, autosomal recessive, with myopathy and facial dysmorphism	< 1 in 500	Reduced

15	MYO1E	Glomerulosclerosis, focal segmental, 6	< 1 in 500	Reduced
10	MYO3A	Deafness, autosomal recessive, type 30	< 1 in 500	Reduced
15	MYO5A	Griscelli syndrome, type 1	< 1 in 500	Reduced
18	MYO5B	Microvillus inclusion disease	< 1 in 500	1 in 25000
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
10	MYPN	Nemaline myopathy, type 11, autosomal recessive	< 1 in 500	Reduced
2	NADK2	2,4-dienoyl-CoA reductase deficiency	1 in 500	Reduced
22	NAGA	Schindler disease, type I; Schindler disease, type III; Kanzaki disease	1 in 500	1 in 5000
17	NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo B)	1 in 346	1 in 1384
17	NAGS	N-acetylglutamate synthase deficiency	< 1 in 500	Reduced
13	NALCN	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	< 1 in 500	Reduced
9	NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type	< 1 in 500	Reduced
11	NARS2	Combined oxidative phosphorylation deficiency 24	< 1 in 500	Reduced
1	NAXE	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy	< 1 in 500	Reduced
2	NBAS	Infantile liver failure syndrome, type 2; Short stature, optic nerve atrophy, and Pelger-Huet anomaly	1 in 500	1 in 7600
3	NBEAL2	Gray platelet syndrome	< 1 in 500	Reduced
8	NBN	Nijmegen breakage syndrome	1 in 525	1 in 17500
11	NCAPD3	Microcephaly 22, primary, autosomal recessive	< 1 in 500	Reduced
7	NCF1	Chronic granulomatous disease, type 1	1 in 343	1 in 1027
1	NCF2	Chronic granulomatous disease, type 2	< 1 in 500	Reduced
22	NCF4	Chronic granulomatous disease, type 3	< 1 in 500	Reduced
16	NDE1	Lissencephaly, type 4 (with microcephaly)	< 1 in 500	Reduced
8	NDRG1	Charcot-Marie-Tooth disease, type 4D	< 1 in 500	Reduced
5	NDST1	Mental retardation, autosomal recessive, type 46	< 1 in 500	Reduced
2	NDUFA10	Mitochondrial complex I deficiency, nuclear type 22	< 1 in 500	Reduced
19	NDUFA11	Mitochondrial complex I deficiency, nuclear type 14	< 1 in 500	Reduced
12	NDUFA12	?Mitochondrial complex I deficiency, nuclear type 23	< 1 in 500	Reduced
5	NDUFA2	Mitochondrial complex I deficiency, nuclear type 13	< 1 in 500	Reduced
12	NDUFA9	Mitochondrial complex I deficiency, nuclear type 26	< 1 in 500	Reduced
15	NDUFAF1	Mitochondrial complex I deficiency, nuclear type 11	< 1 in 500	Reduced
5	NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10	< 1 in 500	Reduced
3	NDUFAF3	Mitochondrial complex I deficiency, nuclear type 18	< 1 in 500	Reduced
20	NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16	1 in 982	1 in 4350
8	NDUFAF6	Mitochondrial complex I deficiency, nuclear type 17	≤ 1 in 500	1 in 16666
2	NDUFB3	Mitochondrial complex I deficiency, nuclear type 25	< 1 in 500	Reduced
8	NDUFB9	Mitochondrial complex I deficiency, nuclear type 24	< 1 in 500	Reduced
2	NDUFS1	Mitochondrial complex I deficiency, nuclear type 5	< 1 in 500	Reduced
1	NDUFS2	Mitochondrial complex I deficiency, nuclear type 6	< 1 in 500	Reduced
11	NDUFS3	Mitochondrial complex I deficiency, nuclear type 8	< 1 in 500	Reduced
5	NDUFS4	Mitochondrial complex I deficiency, nuclear type 1	< 1 in 500	Reduced
5	NDUFS6	Mitochondrial complex I deficiency, nuclear type 9	< 1 in 500	Reduced
19	NDUFS7	Mitochondrial complex I deficiency, nuclear type 3	< 1 in 500	Reduced
11	NDUFS8	Mitochondrial complex I deficiency, nuclear type 2	< 1 in 500	Reduced
11	NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	1 in 469	1 in 736
18	NDUFV2	Mitochondrial complex I deficiency, nuclear type 7	< 1 in 500	Reduced
2	NEB	Nemaline myopathy type 2	1 in 175	1 in 2188
11	NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome; Orofacial cleft 7	< 1 in 500	Reduced
1	NECTIN4	Ectodermal dysplasia-syndactyly syndrome, type 1	< 1 in 500	Reduced
8	NEFL	Charcot-Marie-Tooth disease, type 1F	< 1 in 500	Reduced
4	NEK1	Short-rib thoracic dysplasia, type 6, with or without polydactyly	< 1 in 500	Reduced
17	NEK8	Renal-hepatic-pancreatic dysplasia, type 2	< 1 in 500	Reduced
14	NEK9	Lethal congenital contracture syndrome 10	< 1 in 500	Reduced
6	NEU1	Sialidosis, type 1 and type 2	< 1 in 500	Reduced
10	NEUROG3	Diarrhea 4, malabsorptive, congenital	< 1 in 500	Reduced
2	NFU1	Multiple mitochondrial dysfunctions syndrome 1	< 1 in 500	Reduced
1	NGF	Neuropathy, hereditary sensory and autonomic, type 5	< 1 in 500	Reduced
3	NGLY1	Congenital disorder of deglycosylation	1 in 610	1 in 30500
2	NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	< 1 in 500	Reduced
6	NHLRC1	Epilepsy, progressive myoclonic, type 2B (Lafora)	< 1 in 500	Reduced
5	NHP2	Dyskeratosis congenita, autosomal recessive type 2	1 in 250	1 in 24964
14	NIN	Seckel syndrome, type 7	< 1 in 500	Reduced
5	NIPAL4	Ichthyosis, congenital, autosomal recessive, type 6	< 1 in 500	Reduced
8	NKX2-6	Conotruncal heart malformations	< 1 in 500	Reduced
4	NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia	< 1 in 500	Reduced
10	NKX6-2	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy	< 1 in 500	Reduced
17	NLRP1	Autoinflammation with arthritis and dyskeratosis	< 1 in 500	Reduced
19	NLRP7	Hydatidiform mole, recurrent, type 1	1 in 500	1 in 722
7	NME8	Ciliary dyskinesia, primary, type 6	< 1 in 500	Reduced

1	NMNAT1	Leber congenital amaurosis 9; Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis	< 1 in 500	Reduced
5	NNT	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency	< 1 in 500	Reduced
15	NOP10	Dyskeratosis congenita, autosomal recessive type 1	< 1 in 500	Reduced
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 1393
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
1	NPHS2	Nephrotic syndrome, type 2	1 in 226	1 in 11300
9	NPR2	Acromesomelic dysplasia, Maroteaux type	< 1 in 500	Reduced
12	NR1H4	Cholestasis, progressive familial intrahepatic, type 5	< 1 in 500	Reduced
15	NR2E3	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	1 in 278	1 in 5560
14	NRL	Retinal degeneration, autosomal recessive, clumped pigment type	< 1 in 500	Reduced
2	NRXN1	Pitt-Hopkins-like syndrome, type 2	< 1 in 500	Reduced
8	NSMCE2	Seckel syndrome, type 10	< 1 in 500	Reduced
5	NSUN2	Mental retardation, autosomal recessive, type 5	< 1 in 500	Reduced
10	NT5C2	Spastic paraplegia, type 45, autosomal recessive	< 1 in 500	Reduced
7	NT5C3A	Anemia, hemolytic, due to UMPH1 deficiency	< 1 in 500	Reduced
6	NT5E	Calcification of joints and arteries	< 1 in 500	Reduced
16	NTHL1	Familial adenomatous polyposis, type 3	< 1 in 500	Reduced
1	NTRK1	Insensitivity to pain, congenital, with anhidrosis	1 in 1122	1 in 11220
14	NUBPL	Mitochondrial complex I deficiency, nuclear type 21	1 in 500	1 in 7100
12	NUP107	Nephrotic syndrome, type 11	< 1 in 500	Reduced
19	NUP62	Striatonigral degeneration, infantile	< 1 in 500	Reduced
16	NUP93	Nephrotic syndrome, type 12	< 1 in 500	Reduced
10	OAT	Gyrate atrophy of choroid and retina	< 1 in 500	Reduced
2	OBSL1	3M syndrome 2	1 in 103	1 in 5150
15	OCA2	Oculocutaneous albinism type 2	1 in 101	1 in 1010
5	OCLN	Pseudo-TORCH syndrome, type 1	< 1 in 500	Reduced
19	ODAD1	Ciliary dyskinesia, primary, type 20	< 1 in 500	Reduced
10	ODAD2	Ciliary dyskinesia, primary, type 23	< 1 in 500	Reduced
19	ODAD3	Ciliary dyskinesia, primary, type 30	< 1 in 500	Reduced
3	OPA1	Behr syndrome	1 in 500	1 in 10000
19	OPA3	3-methylglutaconic aciduria, type 3	< 1 in 500	Reduced
10	OPTN	Amyotrophic lateral sclerosis, type 12	< 1 in 500	Reduced
12	ORAI1	Immunodeficiency, type 9	< 1 in 500	Reduced
1	ORC1	Meier-Gorlin syndrome, type 1	< 1 in 500	Reduced
2	ORC4	Meier-Gorlin syndrome, type 2	< 1 in 500	Reduced
16	ORC6	Meier-Gorlin syndrome, type 3	< 1 in 500	Reduced
14	OSGEP	Galloway-Mowat syndrome 3	< 1 in 500	Reduced
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 2270
11	OTOG	Deafness, autosomal recessive, type 18B	< 1 in 500	Reduced
12	OTOGL	Deafness, autosomal recessive, type 84B	< 1 in 500	Reduced
8	OTUD6B	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	< 1 in 500	Reduced
5	OTULIN	Autoinflammation, panniculitis, and dermatosis syndrome	< 1 in 500	Reduced
5	OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency	< 1 in 500	Reduced
3	P2RY12	Bleeding disorder, platelet-type, type 8	< 1 in 500	Reduced
1	P3H1	Osteogenesis imperfecta, type 8	1 in 567	1 in 1447
3	P3H2	Myopia, high, with cataract and vitreoretinal degeneration	< 1 in 500	Reduced
1	PADI6	Preimplantation embryonic lethality 2	< 1 in 500	Reduced
12	PAH	Phenylketonuria	1 in 60	1 in 857
16	PALB2	PALB2-related conditions	< 1 in 500	Reduced
16	PAM16	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type	< 1 in 500	Reduced
20	PANK2	Neurodegeneration with brain iron accumulation type 1	1 in 400	1 in 5000
10	PAPSS2	Brachyolmia, type 4, with mild epiphyseal and metaphyseal changes	< 1 in 500	Reduced
1	PARK7	Parkinson disease, type 7, autosomal recessive, early-onset	< 1 in 500	Reduced
16	PARN	Dyskeratosis congenita, autosomal recessive 6	< 1 in 500	Reduced
15	PATL2	Oocyte maturation defect 4	< 1 in 500	Reduced
1	PAX7	Rhabdomyosarcoma 2, alveolar	< 1 in 500	Reduced
11	PC	Pyruvate carboxylase deficiency	1 in 251	1 in 3150
2	PCARE	Retinitis pigmentosa, type 54	< 1 in 500	Reduced
10	PCBD1	Hyperphenylalaninemia, BH4-deficient, type D	< 1 in 500	Reduced
13	PCCA	Propionic acidemia	1 in 636	1 in 2544
3	PCCB	Propionic acidemia	1 in 635	1 in 7938
5	PCDH12	Microcephaly, seizures, spasticity, and brain calcification	< 1 in 500	Reduced
10	PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	1 in 497	1 in 1657

14	PCK2	PEPCK deficiency, mitochondrial	< 1 in 500	Reduced
21	PCNT	Microcephalic osteodysplastic primordial dwarfism, type 2	< 1 in 500	Reduced
5	PCSK1	Obesity with impaired prohormone processing	< 1 in 500	Reduced
3	PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy	< 1 in 500	Reduced
6	PDE10A	Dyskinesia, limb and orofacial, infantile-onset	< 1 in 500	Reduced
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	< 1 in 500	Reduced
17	PDE6G	Retinitis pigmentosa type 57	< 1 in 500	Reduced
12	PDE6H	Retinal cone dystrophy 3 and achromatopsia 6	< 1 in 500	Reduced
3	PDHB	Pyruvate dehydrogenase E1-beta deficiency	< 1 in 500	Reduced
11	PDHX	Lacticacidemia due to PDX1 deficiency	< 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
21	PDXK	Neuropathy, hereditary motor and sensory, type VIC, with optic atrophy	< 1 in 500	Reduced
10	PDZD7	Deafness, autosomal recessive, type 57; Usher syndrome, type 2C, digenic	< 1 in 500	Reduced
19	PEPD	Prolidase deficiency	1 in 500	1 in 10000
19	PET100	Mitochondrial complex IV deficiency, nuclear type 12	< 1 in 500	Reduced
7	PEX1	Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)	1 in 191	1 in 3820
1	PEX10	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B	1 in 1287	1 in 16088
1	PEX11B	?Peroxisome biogenesis disorder 14B	< 1 in 500	Reduced
17	PEX12	Peroxisome biogenesis disorder type 3A (Zellweger)	< 1 in 500	Reduced
2	PEX13	Peroxisome biogenesis disorder, type 11A (Zellweger syndrome); Peroxisome biogenesis disorder, type 11B	< 1 in 500	Reduced
1	PEX14	Peroxisome biogenesis disorder, type 13A (Zellweger syndrome)	< 1 in 500	Reduced
11	PEX16	Peroxisome biogenesis disorder, type 8A (Zellweger syndrome); Peroxisome biogenesis disorder, type 8B	< 1 in 500	Reduced
1	PEX19	Peroxisome biogenesis disorder, type 12A (Zellweger syndrome)	< 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
6	PEX3	Peroxisome biogenesis disorder, type 10A (Zellweger syndrome)	< 1 in 500	Reduced
12	PEX5	Peroxisome biogenesis disorder type 2A (Zellweger)	< 1 in 500	Reduced
6	PEX6	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	1 in 83	1 in 277
6	PEX7	Rhizomelic chondrodysplasia punctata, type 1	1 in 371	1 in 7420
12	PFKM	Glycogen storage disease, type 7	< 1 in 500	Reduced
7	PGAM2	Glycogen storage disease X	< 1 in 500	Reduced
2	PGAP1	Mental retardation, autosomal recessive 42	< 1 in 500	Reduced
11	PGAP2	Hyperphosphatasia with mental retardation syndrome 3	< 1 in 500	Reduced
17	PGAP3	Hyperphosphatasia with mental retardation syndrome 4	< 1 in 500	Reduced
1	PGM1	Congenital disorder of glycosylation, type 1t	< 1 in 500	Reduced
6	PGM3	Immunodeficiency, type 23	< 1 in 500	Reduced
1	PHGDH	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	1 in 631	1 in 4207
16	PHKB	Glycogen storage disease, type 9B	1 in 450	1 in 9000
16	PHKG2	Glycogen storage disease type 9c	< 1 in 500	Reduced
11	PHOX2A	Fibrosis of extraocular muscles, congenital, 2	< 1 in 500	Reduced
10	PHYH	Refsum disease	< 1 in 500	Reduced
22	PI4KA	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis	< 1 in 500	Reduced
13	PIBF1	Joubert syndrome 33	1 in 500	1 in 10000
16	PIEZO1	Lymphedema, hereditary, type 3	< 1 in 500	Reduced
18	PIEZO2	Arthrogryposis, distal, with impaired proprioception and touch	< 1 in 500	Reduced
1	PIGC	Glycosylphosphatidylinositol biosynthesis defect 16	< 1 in 500	Reduced
4	PIGG	Mental retardation, autosomal recessive 53	< 1 in 500	Reduced
17	PIGL	Zunich neuroectodermal syndrome	< 1 in 500	Reduced
1	PIGM	Glycosylphosphatidylinositol deficiency	< 1 in 500	Reduced
18	PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome, type 1	< 1 in 500	1 in 5000
9	PIGO	Hyperphosphatasia with mental retardation syndrome 2	< 1 in 500	Reduced
20	PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3	< 1 in 500	Reduced
1	PIGV	Hyperphosphatasia with mental retardation syndrome 1	< 1 in 500	Reduced
17	PIGW	Glycosylphosphatidylinositol biosynthesis defect 11	< 1 in 500	Reduced
4	PIGY	Hyperphosphatasia with mental retardation syndrome 6	< 1 in 500	Reduced
1	PINK1	Parkinson disease, type 6, early onset	< 1 in 500	Reduced
19	PIP5K1C	Lethal congenital contractural syndrome, type 3	< 1 in 500	Reduced
2	PJVK	Deafness, autosomal recessive, type 59	< 1 in 500	Reduced
7	PKD1L1	Heterotaxy, visceral, 8, autosomal	< 1 in 500	Reduced
6	PKHD1	Polycystic kidney disease type 4	1 in 66	1 in 264
1	PKLR	Pyruvate kinase deficiency	1 in 160	1 in 3200
1	PKP1	Ectodermal dysplasia/skin fragility syndrome	< 1 in 500	Reduced
22	PLA2G6	Infantile neuroaxonal dystrophy type 1	1 in 343	1 in 856
9	PLAA	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	< 1 in 500	Reduced

20	PLCB1	Epileptic encephalopathy, early infantile, type 12	< 1 in 500	Reduced
20	PLCB4	Auriculocondylar syndrome, type 2	< 1 in 500	Reduced
3	PLCD1	Nail disorder, nonsyndromic congenital, type 3 (leukonychia)	< 1 in 500	Reduced
10	PLCE1	Nephrotic syndrome, type 3	< 1 in 500	Reduced
3	PLD1	Cardiac valvular defect, developmental	< 1 in 500	Reduced
8	PLEC	Epidermolysis bullosa simplex with muscular dystrophy	< 1 in 500	Reduced
1	PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate, type C	< 1 in 500	1 in 16666
6	PLG	Plasminogen deficiency, type I	< 1 in 500	Reduced
4	PLK4	Microcephaly and chorioretinopathy, autosomal recessive, 2	< 1 in 500	Reduced
1	PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	1 in 159	1 in 450
3	PLOD2	Bruck syndrome 2	1 in 1400	1 in 35000
7	PLOD3	Lysyl hydroxylase 3 deficiency	< 1 in 500	Reduced
8	PLPBP	Epilepsy, early-onset, vitamin B6-dependent	< 1 in 500	Reduced
16	PMM2	Congenital disorder of glycosylation, type 1A	1 in 71	1 in 3550
17	PMP22	Dejerine-Sottas disease	< 1 in 500	Reduced
9	PMPCA	Spinocerebellar ataxia, autosomal recessive, type 2	< 1 in 500	Reduced
7	PMPCB	Multiple mitochondrial dysfunctions syndrome 6	< 1 in 500	Reduced
19	PNKP	Microcephaly, seizures, and developmental delay; Ataxia-oculomotor apraxia 4; ?Charcot-Marie-Tooth disease, type 2B2	< 1 in 500	Reduced
14	PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency	< 1 in 500	Reduced
6	PNPLA1	Ichthyosis, congenital, autosomal recessive, type 10	< 1 in 500	Reduced
11	PNPLA2	Neutral lipid storage disease with myopathy	< 1 in 500	Reduced
19	PNPLA6	Boucher-Neuhauser syndrome; Oliver-McFarlane syndrome; Spastic paraplegia, type 39, autosomal recessive	< 1 in 500	Reduced
17	PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	1 in 1107	1 in 3983
2	PNPT1	Combined oxidative phosphorylation deficiency 13	< 1 in 500	Reduced
3	POC1A	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	< 1 in 500	Reduced
12	POC1B	Cone-rod dystrophy 20	< 1 in 500	Reduced
12	POLE	FILS syndrome	< 1 in 500	Reduced
15	POLG	POLG-related disorders	1 in 194	1 in 3800
6	POLH	Xeroderma pigmentosum, variant type	< 1 in 500	Reduced
6	POLR1C	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	1 in 1021	1 in 1659
13	POLR1D	Treacher Collins syndrome, type 2	< 1 in 500	Reduced
10	POLR3A	Leukodystrophy, hypomyelinating, type 7	< 1 in 500	Reduced
12	POLR3B	Leukodystrophy, hypomyelinating, type 8	1 in 500	1 in 16600
2	POMC	Obesity, adrenal insufficiency, and red hair due to POMC deficiency	< 1 in 500	Reduced
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 15750
3	POMGNT2	Muscular dystrophy-dystroglycanopathy, type 8A (Walker-Warburg syndrome); Type 8C (limb-girdle muscular dystrophy, type 24 [LGMD R24])	< 1 in 500	Reduced
8	POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12	< 1 in 500	Reduced
13	POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	< 1 in 500	Reduced
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 3720
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
8	POP1	Anauxetic dysplasia, type 2	< 1 in 500	Reduced
7	POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	1 in 268	1 in 1069
3	POU1F1	Pituitary hormone deficiency, combined, type 1	1 in 1230	Reduced
4	PPA2	Sudden cardiac failure, infantile	< 1 in 500	Reduced
15	PPIB	Osteogenesis imperfecta, type 9	< 1 in 500	Reduced
4	PPM1K	?Maple syrup urine disease, mild variant	1 in 200	1 in 4000
1	PPP1R15B	Microcephaly, short stature, and impaired glucose metabolism 2	< 1 in 500	Reduced
1	PPT1	Ceroid lipofuscinosis, neuronal, type 1	1 in 488	1 in 16200
17	PRCD	Retinitis pigmentosa, type 36	< 1 in 500	Reduced
9	PRDM12	Neuropathy, hereditary sensory and autonomic, type VIII	< 1 in 500	Reduced
4	PRDM5	Brittle cornea syndrome, type 2	< 1 in 500	Reduced
1	PRDX1	Methylmalonic aciduria and homocystinuria, cblC type, digenic	< 1 in 500	Reduced
2	PREPL	Myasthenic syndrome, congenital, type 22	< 1 in 500	Reduced
10	PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2	1 in 308	1 in 538
1	PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	< 1 in 500	Reduced
12	PRICKLE1	Epilepsy, progressive myoclonic, type 1B	< 1 in 500	Reduced
3	PRKCD	Autoimmune lymphoproliferative syndrome, type 3	< 1 in 500	Reduced
6	PRKN	Parkinson disease, type 2, juvenile	< 1 in 500	Reduced
2	PRKRA	Dystonia, type 16	< 1 in 500	Reduced
16	PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures	< 1 in 500	Reduced
2	PROC	Thrombophilia due to protein C deficiency, autosomal recessive	< 1 in 500	Reduced
22	PRODH	Hyperprolinemia, type 1	1 in 500	1 in 3333
4	PROM1	Retinitis pigmentosa, type 41	1 in 323	1 in 6460
5	PROP1	Pituitary hormone deficiency, combined, type 2	1 in 591	1 in 29495
3	PROS1	Thrombophilia due to protein S deficiency, autosomal recessive	1 in 500	1 in 16600
6	PRPH2	Leber congenital amaurosis 18; Retinitis punctata albescens	< 1 in 500	Reduced
1	PRRX1	Agnathia-otocephaly complex	< 1 in 500	Reduced

4	PRSS12	Mental retardation, autosomal recessive, type 1	< 1 in 500	Reduced
2	PRSS56	Microphthalmia, isolated, type 6	< 1 in 500	Reduced
1	PRUNE1	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies	< 1 in 500	Reduced
19	PRX	Charcot-Marie-Tooth disease, type 4F	< 1 in 500	Reduced
10	PSAP	Combined SAP deficiency	< 1 in 500	Reduced
9	PSAT1	Neu-Laxova syndrome, type 2	< 1 in 500	Reduced
6	PSMB8	Autoinflammation, lipodystrophy, and dermatosis syndrome	< 1 in 500	Reduced
17	PSMC3IP	Ovarian dysgenesis 3	< 1 in 500	Reduced
7	PSPH	Phosphoserine phosphatase deficiency	< 1 in 500	Reduced
10	PTF1A	Pancreatic agenesis 2	< 1 in 500	Reduced
11	PTH	Hypoparathyroidism, familial isolated, type 1	< 1 in 500	Reduced
3	PTH1R	Chondrodysplasia, Blomstrand type; Eiken syndrome	< 1 in 500	Reduced
3	PTPN23	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	< 1 in 500	Reduced
1	PTPRC	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	< 1 in 500	Reduced
12	PTPRO	Nephrotic syndrome, type 6	< 1 in 500	Reduced
12	PTPRQ	Deafness, autosomal recessive, type 84A	1 in 500	1 in 3333
17	PTRH2	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease	< 1 in 500	Reduced
11	PTS	Hyperphenylalaninemia, BH4-deficient, type A	1 in 478	1 in 1593
12	PUS1	Myopathy, lactic acidosis, and sideroblastic anemia, type 1	< 1 in 500	Reduced
2	PXDN	Anterior segment dysgenesis, type 7, with sclerocornea	< 1 in 500	Reduced
17	PYCR1	Cutis laxa, autosomal recessive, type 2B	< 1 in 500	Reduced
1	PYCR2	Leukodystrophy, hypomyelinating, type 10	< 1 in 500	Reduced
14	PYGL	Glycogen storage disease, type 6	1 in 500	1 in 10000
11	PYGM	McArdle disease	1 in 206	1 in 2060
12	PYROXD1	Myopathy, myofibrillar, type 8	< 1 in 500	Reduced
3	QARS1	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	< 1 in 500	Reduced
4	QDPR	Hyperphenylalaninemia, BH4-deficient, type C	< 1 in 500	Reduced
10	RAB18	Warburg micro syndrome, type 3	< 1 in 500	Reduced
6	RAB23	Carpenter syndrome	< 1 in 500	Reduced
15	RAB27A	Griscelli syndrome, type 2	< 1 in 500	Reduced
4	RAB28	Cone-rod dystrophy 18	< 1 in 500	Reduced
4	RAB33B	Smith-McCort dysplasia 2	< 1 in 500	Reduced
2	RAB3GAP1	Warburg micro syndrome, type 1	< 1 in 500	Reduced
1	RAB3GAP2	Martsolf syndrome 1; Warburg micro syndrome 2	< 1 in 500	Reduced
5	RAD50	Nijmegen breakage syndrome-like disorder	< 1 in 500	Reduced
17	RAD51C	RAD51C-related conditions	< 1 in 500	Reduced
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 1925	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
3	RARB	Microphthalmia, syndromic 12	< 1 in 500	Reduced
5	RARS1	Leukodystrophy, hypomyelinating, type 9	< 1 in 500	Reduced
6	RARS2	Pontocerebellar hypoplasia, type 6	1 in 269	1 in 3363
15	RASGRP1	Immunodeficiency, type 64	< 1 in 500	Reduced
18	RAX	Isolated microphthalmia, type 3	< 1 in 500	Reduced
18	RBBP8	Jawad syndrome; Seckel syndrome, type 2	< 1 in 500	Reduced
20	RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency	< 1 in 500	Reduced
1	RBM8A	Thrombocytopenia-absent radius syndrome	1 in 500	1 in 1000
10	RBP3	?Retinitis pigmentosa 66	< 1 in 500	Reduced
10	RBP4	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome	< 1 in 500	Reduced
13	RCBTB1	Retinal dystrophy with or without extraocular anomalies	< 1 in 500	Reduced
1	RD3	Leber congenital amaurosis, type 12	< 1 in 500	Reduced
14	RDH12	Leber congenital amaurosis, type 13	1 in 456	1 in 4560
12	RDH5	Fundus albipunctatus	< 1 in 500	Reduced
11	RDX	Deafness, autosomal recessive, type 24	< 1 in 500	Reduced
8	RECQL4	Baller-Gerold syndrome; RAPADILINO syndrome; Rothmund-Thomson syndrome	< 1 in 500	Reduced
19	REEP6	Retinitis pigmentosa 77	< 1 in 500	Reduced
7	RELN	Lissencephaly 2 (Norman-Roberts type)	< 1 in 500	Reduced
1	REN	Renal tubular dysgenesis	< 1 in 500	Reduced
5	RETREG1	Neuropathy, hereditary sensory and autonomic, type 2B	< 1 in 500	Reduced
3	RFT1	Congenital disorder of glycosylation, type In	< 1 in 500	Reduced
1	RFX5	Bare lymphocyte syndrome, type 2	< 1 in 500	Reduced
6	RFX6	Mitchell-Riley syndrome	< 1 in 500	Reduced
19	RFXANK	Bare lymphocyte syndrome, type 2, complementation group B	< 1 in 500	Reduced
13	RFXAP	Bare lymphocyte syndrome, type 2	< 1 in 500	Reduced
3	RHO	Retinitis pigmentosa, type 4; Retinitis punctata albescens	1 in 416	1 in 8320
20	RIN2	Macs syndrome	< 1 in 500	Reduced
21	RIPK4	Popliteal pterygium syndrome, Bartsocas-Papas type	< 1 in 500	Reduced
6	RIPOR2	Deafness, autosomal recessive, type 104	< 1 in 500	Reduced
15	RLBP1	Bothnia retinal dystrophy; Fundus albipunctatus	< 1 in 500	Reduced

6	RMND1	Combined oxidative phosphorylation deficiency 11	< 1 in 500	Reduced
9	RMRP	Anauxetic dysplasia, type 1	1 in 500	1 in 6250
2	RNASEH1	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2	< 1 in 500	Reduced
19	RNASEH2A	Aicardi-Goutieres syndrome, type 4	< 1 in 500	Reduced
13	RNASEH2B	Aicardi-Goutieres syndrome, type 2	1 in 440	1 in 7333
11	RNASEH2C	Aicardi-Goutieres syndrome, type 3	< 1 in 500	Reduced
6	RNASET2	Leukoencephalopathy, cystic, without megalencephaly	< 1 in 500	Reduced
3	RNF168	RIDDLE syndrome	< 1 in 500	Reduced
7	RNF216	Gordon Holmes syndrome	< 1 in 500	Reduced
11	ROBO3	Gaze palsy, familial horizontal, with progressive scoliosis, type 1	< 1 in 500	Reduced
16	ROGDI	Kohlschutter-Tonz syndrome	< 1 in 500	Reduced
11	ROM1	Retinitis pigmentosa, type 7, digenic	< 1 in 500	Reduced
9	ROR2	Robinow syndrome, autosomal recessive	< 1 in 500	Reduced
1	RORC	Immunodeficiency, type 42	< 1 in 500	Reduced
8	RP1	Retinitis pigmentosa, type 1	< 1 in 500	Reduced
1	RPE65	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy	1 in 366	1 in 18300
14	RPGRIP1	Leber congenital amaurosis, type 6	< 1 in 500	Reduced
16	RPGRIP1L	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	1 in 319	1 in 860
8	RRM2B	Mitochondrial DNA depletion syndrome, type 8A (encephalomyopathic type with renal tubulopathy) and type 8B (MNGIE type)	< 1 in 500	Reduced
21	RSPH1	Ciliary dyskinesia, primary, type 24	< 1 in 500	Reduced
6	RSPH3	Ciliary dyskinesia, primary, type 32	< 1 in 500	Reduced
6	RSPH4A	Ciliary dyskinesia, primary, type 11	< 1 in 500	Reduced
6	RSPH9	Ciliary dyskinesia, primary, type 12	< 1 in 500	Reduced
20	RSPO4	Anonychia congenita	< 1 in 500	Reduced
16	RSPRY1	Spondyloepimetaphyseal dysplasia, Faden-Alkuraya type	< 1 in 500	Reduced
20	RTEL1	Dyskeratosis congenita, autosomal recessive type 5	< 1 in 500	Reduced
6	RTN4IP1	Optic atrophy 10 with or without ataxia, mental retardation, and seizures	< 1 in 500	Reduced
18	RTTN	Microcephaly, short stature, and polymicrogyria with seizures	< 1 in 500	Reduced
9	RUSC2	Mental retardation, autosomal recessive 61	< 1 in 500	Reduced
12	RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	< 1 in 500	Reduced
19	RYR1	Minicore myopathy with external ophthalmoplegia	< 1 in 500	1 in 10000
19	S1PR2	Deafness, autosomal recessive, type 68	< 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	SAMD9	Tumoral calcinosis, familial, normophosphatemic	< 1 in 500	Reduced
20	SAMHD1	Aicardi-Goutieres syndrome, type 5	1 in 610	1 in 2033
5	SAR1B	Chylomicron retention disease	< 1 in 500	Reduced
19	SARS2	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	< 1 in 500	Reduced
7	SBDS	Shwachman-Diamond syndrome	1 in 224	1 in 804
22	SBF1	Charcot-Marie-Tooth disease, type 4B3	< 1 in 500	Reduced
11	SBF2	Charcot-Marie-Tooth disease, type 4B2	< 1 in 500	Reduced
11	SC5D	Lathosterolosis	< 1 in 500	Reduced
4	SCARB2	Epilepsy, progressive myoclonic, type 4, with or without renal failure	< 1 in 500	Reduced
22	SCARF2	Van den Ende-Gupta syndrome	< 1 in 500	Reduced
19	SCN1B	Epileptic encephalopathy, early infantile, type 52	< 1 in 500	Reduced
17	SCN4A	Myasthenic syndrome, congenital, type 16	< 1 in 500	Reduced
2	SCN9A	Indifference to pain and autosomal recessive hereditary sensory neuropathy type 2D	< 1 in 500	Reduced
12	SCNN1A	Pseudohypoaldosteronism, type 1	< 1 in 500	Reduced
16	SCNN1B	Pseudohypoaldosteronism, type 1	< 1 in 500	Reduced
16	SCNN1G	Pseudohypoaldosteronism, type 1	< 1 in 500	Reduced
17	SCO1	Mitochondrial complex IV deficiency, nuclear type 4	< 1 in 500	Reduced
22	SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1	1 in 500	1 in 8333
11	SCYL1	Spinocerebellar ataxia, autosomal recessive, type 21	< 1 in 500	Reduced
1	SDCCAG8	Bardet-Biedl syndrome, type 16	< 1 in 500	Reduced
5	SDHA	Mitochondrial respiratory chain complex II deficiency; Leigh syndrome	< 1 in 500	Reduced
19	SDHAF1	Mitochondrial complex II deficiency	< 1 in 500	Reduced
12	SDR9C7	Ichthyosis, congenital, autosomal recessive 13	< 1 in 500	1 in 16666
14	SEC23A	Craniolenticulosutural dysplasia	< 1 in 500	Reduced
20	SEC23B	Dyserythropoietic anemia, congenital, type 2	1 in 500	1 in 16600
4	SEC24D	Cole-Carpenter syndrome 2	< 1 in 500	Reduced
9	SECISBP2	Thyroid hormone metabolism, abnormal	< 1 in 500	Reduced
1	SELENON	Muscular dystrophy, rigid spine, type 1	< 1 in 500	Reduced
1	SEMA4A	Cone-rod dystrophy, type 10; Retinitis pigmentosa, type 35	< 1 in 500	Reduced
4	SEPSECS	Pontocerebellar hypoplasia, type 2D	< 1 in 500	Reduced
6	SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome (MEGDEL)	< 1 in 500	Reduced
14	SERPINA1	Alpha-1 antitrypsin deficiency	1 in 24	1 in 2400
18	SERPINB7	Palmoplantar keratoderma, Nagashima type	< 1 in 500	Reduced
18	SERPINB8	Peeling skin syndrome 5	< 1 in 500	Reduced
1	SERPINC1	Thrombophilia due to antithrombin III deficiency	< 1 in 500	Reduced

7	SERPINE1	Plasminogen activator inhibitor-1 deficiency	< 1 in 500	Reduced
17	SERPINF1	Osteogenesis imperfecta, type 6	< 1 in 500	Reduced
17	SERPINF2	Alpha-2-plasmin inhibitor deficiency	< 1 in 500	Reduced
11	SERPING1	Angioedema, hereditary, types 1 and 2	< 1 in 500	Reduced
11	SERPINH1	Osteogenesis imperfecta, type 10	< 1 in 500	Reduced
9	SETX	Spinocerebellar ataxia, autosomal recessive, type 1	1 in 500	1 in 2273
7	SFRP4	Pyle disease	< 1 in 500	Reduced
2	SFTPB	Surfactant metabolism dysfunction, pulmonary, type 1	< 1 in 500	Reduced
10	SFXN4	Combined oxidative phosphorylation deficiency 18	< 1 in 500	Reduced
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
5	SGCD	Limb-girdle muscular dystrophy, type 6 (LGMD R6)	< 1 in 500	Reduced
13	SGCG	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	1 in 1132	1 in 5468
10	SGPL1	Nephrotic syndrome, type 14	< 1 in 500	Reduced
17	SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo A)	1 in 253	1 in 5060
5	SH3PXD2B	Frank-ter Haar syndrome	< 1 in 500	Reduced
5	SH3TC2	Charcot-Marie-Tooth disease, type 4C	1 in 130	1 in 1300
3	SI	Sucrase-isomaltase deficiency, congenital	< 1 in 500	1 in 10000
5	SIL1	Marinesco-Sjogren syndrome	< 1 in 500	Reduced
14	SIX6	Optic disc anomalies with retinal and/or macular dystrophy	< 1 in 500	Reduced
6	SKIC2	Trichohepatoenteric syndrome, type 2 (diarrhea, syndromic)	1 in 500	1 in 10000
5	SKIC3	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	1 in 640	1 in 64000
13	SLC10A2	Bile acid malabsorption, primary	< 1 in 500	Reduced
12	SLC11A2	Anemia, hypochromic microcytic, with iron overload 1	< 1 in 500	Reduced
15	SLC12A1	Bartter syndrome, type 1	< 1 in 500	Reduced
16	SLC12A3	Gitelman syndrome	1 in 73	1 in 2400
20	SLC12A5	Epileptic encephalopathy, early infantile, 34	< 1 in 500	Reduced
15	SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	< 1 in 500	Reduced
17	SLC13A5	Epileptic encephalopathy, early infantile, 25	< 1 in 500	Reduced
1	SLC16A1	Monocarboxylate transporter 1 deficiency	< 1 in 500	Reduced
6	SLC17A5	Salla disease	1 in 328	1 in 2187
10	SLC18A3	Myasthenic syndrome, congenital, 21, presynaptic	< 1 in 500	Reduced
1	SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	1 in 500	1 in 888
2	SLC19A3	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)	1 in 232	1 in 1785
9	SLC1A1	Dicarboxylic aminoaciduria	< 1 in 500	Reduced
2	SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	< 1 in 500	1 in 25000
11	SLC22A12	Hypouricemia, renal	< 1 in 500	Reduced
5	SLC22A5	Carnitine deficiency, systemic primary	1 in 251	1 in 717
15	SLC24A1	Night blindness, congenital stationary (complete), type 1D, autosomal recessive	< 1 in 500	Reduced
14	SLC24A4	Amelogenesis imperfecta, type IIA5	< 1 in 500	Reduced
15	SLC24A5	Albinism, oculocutaneous, type 6	< 1 in 500	Reduced
22	SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria	< 1 in 500	Reduced
2	SLC25A12	Epileptic encephalopathy, early infantile, type 39	< 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
17	SLC25A19	Microcephaly, Amish type; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)	< 1 in 500	Reduced
3	SLC25A20	Carnitine-acylcarnitine translocase deficiency	< 1 in 500	Reduced
11	SLC25A22	Epileptic encephalopathy, early infantile, type 3	< 1 in 500	Reduced
3	SLC25A26	Combined oxidative phosphorylation deficiency 28	< 1 in 500	Reduced
12	SLC25A3	Mitochondrial phosphate carrier deficiency	< 1 in 500	Reduced
3	SLC25A38	Anemia, sideroblastic, type 2, pyridoxine-refractory	< 1 in 500	Reduced
4	SLC25A4	Mitochondrial DNA depletion syndrome, type 12B (cardiomyopathic type) AR	< 1 in 500	Reduced
5	SLC25A46	Neuropathy, hereditary motor and sensory, type VIB	< 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	SLC26A3	Diarrhea 1, secretory chloride, congenital	< 1 in 500	Reduced
7	SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome	1 in 88	1 in 587
7	SLC26A5	?Deafness, autosomal recessive, type 61	< 1 in 500	Reduced
9	SLC27A4	Ichthyosis prematurity syndrome	< 1 in 500	Reduced
10	SLC29A3	Histiocytosis-lymphadenopathy plus syndrome	< 1 in 500	Reduced
1	SLC2A1	GLUT1 deficiency syndrome 1, infantile onset, severe	< 1 in 500	Reduced
20	SLC2A10	Arterial tortuosity syndrome	1 in 1113	1 in 55650
3	SLC2A2	Fanconi-Bickel syndrome	< 1 in 500	Reduced
4	SLC2A9	Hypouricemia, renal, type 2	< 1 in 500	Reduced
1	SLC30A10	Hypermanganesemia with dystonia, type 1	< 1 in 500	Reduced
3	SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration	< 1 in 500	Reduced
5	SLC34A1	Hypercalcemia, infantile, type 2	< 1 in 500	Reduced
4	SLC34A2	Pulmonary alveolar microlithiasis	< 1 in 500	Reduced
9	SLC34A3	Hypophosphatemic rickets with hypercalciuria	< 1 in 500	Reduced
6	SLC35A1	Congenital disorder of glycosylation, type 2F	< 1 in 500	<1 in 1500

1	SLC35A3	Arthrogryposis, impaired intellectual development, and seizures	< 1 in 500	Reduced
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
16	SLC38A8	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis	< 1 in 500	1 in 10000
11	SLC39A13	Ehlers-Danlos syndrome, spondylodysplastic type, 3	< 1 in 500	Reduced
8	SLC39A14	Hypermanganesemia with dystonia 2	< 1 in 500	Reduced
8	SLC39A4	Acrodermatitis enteropathica	1 in 316	1 in 1580
4	SLC39A8	Congenital disorder of glycosylation, type IIn	< 1 in 500	Reduced
2	SLC3A1	Cystinuria	1 in 42	1 in 1050
1	SLC45A1	Intellectual developmental disorder with neuropsychiatric features	< 1 in 500	Reduced
5	SLC45A2	Albinism, oculocutaneous, type 4	< 1 in 500	<1 in 1600
17	SLC46A1	Folate malabsorption, hereditary	< 1 in 500	Reduced
17	SLC4A1	Distal renal tubular acidosis	1 in 500	1 in 10000
20	SLC4A11	Corneal endothelial dystrophy, autosomal recessive	1 in 806	1 in 26800
4	SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities	< 1 in 500	Reduced
8	SLC52A2	Brown-Vialetto-Van Laere syndrome, type 2	< 1 in 500	Reduced
20	SLC52A3	Brown-Vialetto-Van Laere syndrome, type 1	< 1 in 500	Reduced
22	SLC5A1	Glucose/galactose malabsorption	< 1 in 500	Reduced
16	SLC5A2	Renal glucosuria	< 1 in 500	1 in 7143
19	SLC5A5	Thyroid dyshormonogenesis, type 1	< 1 in 500	Reduced
2	SLC5A7	Myasthenic syndrome, congenital, type 20, presynaptic	< 1 in 500	Reduced
1	SLC6A17	Mental retardation, autosomal recessive 48	< 1 in 500	Reduced
5	SLC6A19	Hartnup disorder	1 in 87	1 in 124
5	SLC6A3	Parkinsonism-dystonia, infantile	< 1 in 500	Reduced
11	SLC6A5	Hyperekplexia, type 3	< 1 in 500	Reduced
1	SLC6A9	Glycine encephalopathy with normal serum glycine	< 1 in 500	Reduced
3	SLC7A14	Retinitis pigmentosa 68	< 1 in 500	Reduced
14	SLC7A7	Lysinuric protein intolerance	< 1 in 500	Reduced
19	SLC7A9	Cystinuria	1 in 42	1 in 2100
5	SLC9A3	Diarrhea 8, secretory sodium, congenital	< 1 in 500	Reduced
3	SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive, type 2	< 1 in 500	Reduced
13	SLITRK6	Deafness and myopia	< 1 in 500	Reduced
8	SLURP1	Meleda disease	< 1 in 500	Reduced
16	SLX4	Fanconi anemia, complementation group P	< 1 in 500	Reduced
2	SMARCAL1	Schimke immunoosseous dysplasia	1 in 451	1 in 3007
17	SMARCD2	Specific granule deficiency 2	< 1 in 500	Reduced
19	SMG9	Heart and brain malformation syndrome	< 1 in 500	Reduced
5	SMN1	Spinal muscular atrophy	1 in 50	1 in 588
14	SMOC1	Microphthalmia. with limb anomalies	< 1 in 500	Reduced
6	SMOC2	Dentin dysplasia, type 1, with microdontia and misshapen teeth	< 1 in 500	Reduced
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
7	SNX10	Osteopetrosis, autosomal recessive, type 8	< 1 in 500	Reduced
6	SNX14	Spinocerebellar ataxia, autosomal recessive, type 20	< 1 in 500	Reduced
6	SOBP	Mental retardation, anterior maxillary protrusion, and strabismus	< 1 in 500	Reduced
21	SOD1	Spastic tetraplegia and axial hypotonia, progressive; Amyotrophic lateral sclerosis, type 1	< 1 in 500	Reduced
9	SOHLH1	Ovarian dysgenesis 5	< 1 in 500	Reduced
17	SOST	Sclerosteosis, type 1; Van Buchem disease	< 1 in 500	Reduced
20	SOX18	Hypotrichosis-lymphedema-telangiectasia syndrome	< 1 in 500	Reduced
2	SP110	Hepatic venoocclusive disease with immunodeficiency	< 1 in 500	Reduced
12	SP7	Osteogenesis imperfecta, type XII	< 1 in 500	Reduced
8	SPAG1	Ciliary dyskinesia, primary, type 28	< 1 in 500	Reduced
5	SPARC	Osteogenesis imperfecta, type XVII	< 1 in 500	Reduced
13	SPART	Spactic paraplegia, type 20, autosomal recessive	< 1 in 500	Reduced
14	SPATA7	Leber congenital amaurosis, type 3	< 1 in 500	Reduced
2	SPEG	Centronuclear myopathy, type 5	< 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
15	SPG21	Mast syndrome	< 1 in 500	Reduced
16	SPG7	Spastic paraplegia, type 7, autosomal recessive	1 in 80	1 in 1600
5	SPINK1	Tropical calcific pancreatitis	< 1 in 500	Reduced
5	SPINK5	Netherton syndrome	1 in 500	1 in 25000
19	SPINT2	Diarrhea 3, secretory sodium, congenital, syndromic	< 1 in 500	Reduced
2	SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	< 1 in 500	Reduced
1	SPRTN	Ruijs-Aalfs syndrome	< 1 in 500	Reduced
1	SPTA1	Pyropoikilocytosis; Spherocytosis, type 3	< 1 in 500	Reduced
11	SPTBN2	Spinocerebellar ataxia, autosomal recessive, type 14	< 1 in 500	Reduced
19	SPTBN4	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness	< 1 in 500	Reduced
5	SQSTM1	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	< 1 in 500	Reduced

2	SRD5A2	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)	1 in 400	1 in 4000
4	SRD5A3	Congenital disorder of glycosylation, type 1Q; Kahrizi syndrome	< 1 in 500	Reduced
11	ST14	Ichthyosis, congenital, autosomal recessive, type 11	< 1 in 500	Reduced
1	ST3GAL3	Mental retardation, autosomal recessive 12	< 1 in 500	Reduced
2	ST3GAL5	Salt and pepper developmental regression syndrome	< 1 in 500	Reduced
12	STAC3	Native American myopathy	< 1 in 500	Reduced
7	STAG3	Premature ovarian failure, type 8; Spermatogenic failure 61	< 1 in 500	Reduced
2	STAMBP	Microcephaly-capillary malformation syndrome	< 1 in 500	Reduced
8	STAR	Lipoid adrenal hyperplasia	1 in 1147	1 in 14338
2	STAT1	Immunodeficiency, type 31B, mycobacterial and viral infections	< 1 in 500	Reduced
12	STAT2	Immunodeficiency, type 44	< 1 in 500	Reduced
17	STAT5B	Laron syndrome with immunodeficiency	< 1 in 500	Reduced
1	STIL	Microcephaly, type 7, primary, autosomal recessive	< 1 in 500	Reduced
11	STIM1	Immunodeficiency, type 10	< 1 in 500	Reduced
20	STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	< 1 in 500	Reduced
15	STRA6	Microphthalmia, isolated, with coloboma, type 8	< 1 in 500	Reduced
17	STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy	< 1 in 500	Reduced
15	STRC	Deafness, autosomal recessive, type 16	1 in 68	1 in 80
16	STUB1	Spinocerebellar ataxia, autosomal recessive, type 16	< 1 in 500	Reduced
6	STX11	Hemophagocytic lymphohistiocytosis, familial, type 4	< 1 in 500	Reduced
19	STXBP2	Hemophagocytic lymphohistiocytosis, familial, type 5	< 1 in 500	Reduced
13	SUCLA2	Mitochondrial DNA depletion syndrome, type 5 (encephalomyopathic with or without methylmalonic aciduria)	< 1 in 500	Reduced
2	SUCLG1	Mitochondrial DNA depletion syndrome, type 9 (encephalomyopathic, type with methylmalonic aciduria)	< 1 in 500	Reduced
10	SUFU	Joubert syndrome, type 32	< 1 in 500	Reduced
7	SUGCT	Glutaric aciduria, type 3	< 1 in 500	Reduced
19	SULT2B1	Ichthyosis, congenital, autosomal recessive, type 14	< 1 in 500	Reduced
3	SUMF1	Multiple sulfatase deficiency	< 1 in 500	Reduced
20	SUN5	Spermatogenic failure, type 16	< 1 in 500	Reduced
12	SUOX	Sulfite oxidase deficiency	< 1 in 500	Reduced
9	SURF1	Mitochondrial complex IV deficiency, nuclear type 1; Charcot-Marie-Tooth disease, type 4K	1 in 191	1 in 329
6	SYNE1	Spinocerebellar ataxia, autosomal recessive, type 8	< 1 in 500	Reduced
19	SYNE4	Deafness, autosomal recessive, type 76	1 in 500	1 in 16600
21	SYNJ1	Epileptic encephalopathy, early infantile, 53	< 1 in 500	Reduced
1	SYT14	?Spinocerebellar ataxia, autosomal recessive, type 11	< 1 in 500	Reduced
1	SZT2	Epileptic encephalopathy, early infantile, 18	< 1 in 500	1 in 16666
12	TAC3	Hypogonadotropic hypogonadism, type 10, with or without anosmia	< 1 in 500	Reduced
17	TACO1	Mitochondrial complex IV deficiency, nuclear type 8	< 1 in 500	Reduced
4	TACR3	Hypogonadotropic hypogonadism, type 11, with or without anosmia	< 1 in 500	Reduced
1	TACSTD2	Corneal dystrophy, gelatinous drop-like	< 1 in 500	Reduced
1	TAF13	Mental retardation, autosomal recessive 60	< 1 in 500	Reduced
8	TAF2	Mental retardation, autosomal recessive 40	< 1 in 500	Reduced
7	TAF6	Alazami-Yuan syndrome	< 1 in 500	Reduced
11	TALDO1	Transaldolase deficiency	< 1 in 500	Reduced
22	TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	1 in 566	1 in 1415
6	TAP1	Bare lymphocyte syndrome, type 1	< 1 in 500	Reduced
6	TAP2	Bare lymphocyte syndrome, type 1, due to TAP2 deficiency	< 1 in 500	Reduced
6	TAPBP	Bare lymphocyte syndrome, type 1	< 1 in 500	Reduced
4	TAPT1	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type	< 1 in 500	Reduced
16	TAT	Tyrosinemia, type 2	< 1 in 500	Reduced
20	TBC1D20	Warburg micro syndrome 4	< 1 in 500	Reduced
3	TBC1D23	Pontocerebellar hypoplasia, type 11	< 1 in 500	Reduced
16	TBC1D24	DOORS (deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures) syndrome; Epileptic encephalopathy, early infantile, type 16; Deafness, autosomal recessive, type 86	< 1 in 500	Reduced
6	TBC1D7	Macrocephaly/megalencephaly syndrome, autosomal recessive	< 1 in 500	Reduced
17	TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	< 1 in 500	1 in 5000
1	TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy; Hypoparathyroidism-retardation-dysmorphism syndrome; Kenny-Caffey syndrome, type 1	< 1 in 500	Reduced
4	TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	< 1 in 500	Reduced
1	TBX15	Cousin syndrome	< 1 in 500	Reduced
1	TBX19	Congenital isolated adrenocorticotropic hormone deficiency	< 1 in 500	Reduced
7	TBXAS1	Ghosal syndrome	< 1 in 500	Reduced
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
22	TCN2	Transcobalamin II deficiency	< 1 in 500	Reduced
12	TCTN1	Joubert syndrome, type 13	< 1 in 500	Reduced
12	TCTN2	Joubert syndrome, type 24; ?Meckel syndrome, type 8	< 1 in 500	Reduced
10	TCTN3	Joubert syndrome 18; Orofaciodigital syndrome IV	< 1 in 500	Reduced
14	TDP1	?Spinocerebellar ataxia, autosomal recessive with axonal neuropathy	< 1 in 500	Reduced
6	TDP2	Spinocerebellar ataxia, autosomal recessive, type 23	< 1 in 500	Reduced
9	TDRD7	Cataract 36	< 1 in 500	Reduced

14	TECPR2	Spastic paraplegia, type 49, autosomal recessive	1 in 1946	1 in 12973
19	TECR	Mental retardation, autosomal recessive, type 14	< 1 in 500	Reduced
4	TECRL	Ventricular tachycardia, catecholaminergic polymorphic, 3	< 1 in 500	Reduced
11	TECTA	Deafness, autosomal recessive, type 21	< 1 in 500	Reduced
16	TELO2	You-Hoover-Fong syndrome	< 1 in 500	Reduced
4	TENM3	Microphthalmia, isolated, with coloboma 9	< 1 in 500	Reduced
5	TERT	Dyskeratosis congenita, autosomal recessive, type 4	< 1 in 500	Reduced
8	TEX15	Spermatogenic failure, type 25	< 1 in 500	Reduced
3	TF	Atransferrinemia	1 in 500	1 in 7143
7	TFR2	Hemochromatosis, type 3	< 1 in 500	Reduced
3	TFRC	Immunodeficiency, type 46	< 1 in 500	Reduced
8	TG	Thyroid dyshormonogenesis, type 3	1 in 159	1 in 268
13	TGDS	Catel-Manzke syndrome	< 1 in 500	Reduced
14	TGM1	Ichthyosis, congenital, autosomal recessive, type 1	1 in 186	1 in 1860
15	TGM5	Peeling skin syndrome, type 2	< 1 in 500	Reduced
11	TH	Segawa syndrome, recessive	< 1 in 500	Reduced
16	THOC6	Beaulieu-Boycott-Innes syndrome	< 1 in 500	Reduced
3	THRB	Thyroid hormone resistance, autosomal recessive	< 1 in 500	Reduced
19	TIMM50	3-methylglutaconic aciduria, type 9	< 1 in 500	Reduced
3	TIMMDC1	Mitochondrial complex I deficiency, nuclear type 31	< 1 in 500	Reduced
9	TJP2	Cholestasis, progressive familial intrahepatic 4; Hypercholanemia, familial 1	< 1 in 500	Reduced
16	TK2	Mitochondrial DNA depletion syndrome , type 2 (myopathic type)	1 in 500	1 in 16667
3	TKT	Short stature, developmental delay, and congenital heart defects	< 1 in 500	Reduced
19	TLE6	Preimplantation embryonic lethality	< 1 in 500	Reduced
9	TMC1	Deafness, autosomal recessive, type 7	1 in 400	1 in 20000
17	TMC6	Epidermodysplasia verruciformis	< 1 in 500	Reduced
17	TMC8	Epidermodysplasia verruciformis	< 1 in 500	Reduced
1	TMCO1	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	< 1 in 500	Reduced
17	TMEM107	Meckel syndrome, type 13; Orofaciodigital syndrome, type 16	< 1 in 500	Reduced
11	TMEM126A	Optic atrophy 7	< 1 in 500	Reduced
11	TMEM126B	Mitochondrial complex I deficiency, nuclear type 29	< 1 in 500	Reduced
11	TMEM138	Joubert syndrome 16	< 1 in 500	Reduced
4	TMEM165	Congenital disorder of glycosylation, type 2K	< 1 in 500	Reduced
17	TMEM199	Congenital disorder of glycosylation, type 2P	< 1 in 500	Reduced
11	TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2	< 1 in 500	Reduced
16	TMEM231	Joubert syndrome, type 20; Meckel syndrome,type 11	1 in 560	1 in 18600
2	TMEM237	Joubert syndrome, type 14	< 1 in 500	Reduced
14	TMEM260	Structural heart defects and renal anomalies syndrome	< 1 in 500	Reduced
8	TMEM67	Meckel syndrome 3; COACH syndrome 1; Joubert syndrome 6; Nephronophthisis 11	1 in 147	1 in 2940
8	TMEM70	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	< 1 in 500	Reduced
3	TMIE	Deafness, autosomal recessive, type 6	< 1 in 500	Reduced
21	TMPRSS15	Enterokinase deficiency	< 1 in 500	Reduced
21	TMPRSS3	Deafness, autosomal recessive, type 8/10	1 in 135	1 in 2700
22	TMPRSS6	Iron-refractory iron deficiency anemia	< 1 in 500	Reduced
12	TMTC3	Lissencephaly 8	< 1 in 500	Reduced
18	TNFRSF11A	Osteopetrosis, autosomal recessive, type 7	< 1 in 500	Reduced
8	TNFRSF11B	Paget disease of bone, type 5, juvenile-onset	< 1 in 500	Reduced
17	TNFRSF13B	Immunodeficiency, common variable, type 2	1 in 500	1 in 10000
13	TNFSF11	Osteopetrosis, autosomal recessive, type 2	< 1 in 500	Reduced
3	TNIK	Mental retardation, autosomal recessive 54	< 1 in 500	Reduced
19	TNNT1	Nemaline myopathy , type 5, Amish type	< 1 in 500	Reduced
6	TNXB	Ehlers-Danlos syndrome, classic-like	1 in 335	1 in 1675
1	TOE1	Pontocerebellar hypoplasia, type 7	< 1 in 500	Reduced
17	TOP3A	Microcephaly, growth restriction, and increased sister chromatid exchange 2	< 1 in 500	Reduced
20	TP53RK	Galloway-Mowat syndrome 4	< 1 in 500	Reduced
12	TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency	< 1 in 500	Reduced
7	TPK1	Episodic encephalopathy due to thiamine pyrophosphokinase deficiency	< 1 in 500	Reduced
1	TPM3	Nemaline myopathy, type 1; Congenital fiber-type disproportion myopathy	< 1 in 500	Reduced
2	TPO	Thyroid dyshormonogenesis, type 2A	1 in 72	1 in 2400
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	< 1 in 500	Reduced
2	TRAF3IP1	Senior-Loken syndrome, type 9	< 1 in 500	Reduced
3	TRAIP	Seckel syndrome, type 9	< 1 in 500	Reduced
4	TRAPPC11	Limb-girdle muscular dystrophy, type 18 (LGMD R18)	< 1 in 500	Reduced
2	TRAPPC12	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity	< 1 in 500	Reduced
14	TRAPPC6B	Neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy	< 1 in 500	Reduced
8	TRAPPC9	Mental retardation, autosomal recessive, type 13	< 1 in 500	Reduced
6	TRDN	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	1 in 287	Reduced
6	TREM2	Nasu-Hakola disease	< 1 in 500	Reduced

3	TREX1	Aicardi-Goutieres syndrome, type 1	1 in 500	1 in 10000
8	TRHR	Hypothyroidism, congenital, nongoitrous, type 7	< 1 in 500	Reduced
4	TRIM2	Charcot-Marie-Tooth disease, type 2R	< 1 in 500	Reduced
9	TRIM32	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	1 in 1273	1 in 12730
17	TRIM37	Mulibrey nanism	< 1 in 500	Reduced
22	TRIOBP	Deafness, autosomal recessive, type 28	1 in 445	1 in 8900
14	TRIP11	Achondrogenesis, type 1A	< 1 in 500	Reduced
5	TRIP13	Mosaic variegated aneuploidy syndrome 3	< 1 in 500	Reduced
15	TRIP4	Spinal muscular atrophy with congenital bone fractures 1	< 1 in 500	Reduced
1	TRIT1	Combined oxidative phosphorylation deficiency 35	< 1 in 500	Reduced
4	TRMT10A	Microcephaly, short stature, and impaired glucose metabolism 1	< 1 in 500	Reduced
3	TRMT10C	Combined oxidative phosphorylation deficiency 30	< 1 in 500	Reduced
14	TRMT5	Combined oxidative phosphorylation deficiency 26	< 1 in 500	Reduced
22	TRMU	Liver failure, transient infantile	< 1 in 500	Reduced
3	TRNT1	Retinitis pigmentosa and erythrocytic microcytosis	< 1 in 500	Reduced
15	TRPM1	Night blindness, congenital stationary (complete), type 1C, autosomal recessive	1 in 500	1 in 10000
9	TRPM6	Familial hypomagnesemia with secondary hypocalcemia	< 1 in 500	Reduced
7	TRPV6	Hyperparathyroidism, transient neonatal	< 1 in 500	Reduced
1	TSEN15	Pontocerebellar hypoplasia, type 2F	< 1 in 500	Reduced
3	TSEN2	Pontocerebellar hypoplasia, type 2B	< 1 in 500	Reduced
19	TSEN34	Pontocerebellar hypoplasia type 2C	< 1 in 500	Reduced
17	TSEN54	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	1 in 223	1 in 3997
12	TSFM	Combined oxidative phosphorylation deficiency, type 3	< 1 in 500	Reduced
1	TSHB	Hypothyroidism, congenital, nongoitrous, type 4	< 1 in 500	Reduced
14	TSHR	Hypothyroidism, congenital, nongoitrous, type 1	1 in 325	1 in 3250
17	TTC19	Mitochondrial complex III deficiency, nuclear type 2	< 1 in 500	Reduced
2	TTC21B	Short-rib thoracic dysplasia, type 4, with or without polydactyly	< 1 in 500	Reduced
2	TTC7A	Gastrointestinal defects and immunodeficiency syndrome	< 1 in 500	Reduced
14	TTC8	Bardet-Biedl syndrome, type 8	< 1 in 500	Reduced
8	TTI2	Mental retardation, autosomal recessive, type 39	< 1 in 500	Reduced
14	TTLL5	Cone-rod dystrophy 19	< 1 in 500	Reduced
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
22	TUBA8	Cortical dysplasia, complex, with other brain malformations, type 8	< 1 in 500	Reduced
15	TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, type 3	1 in 500	1 in 10000
22	TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, type 1	< 1 in 500	Reduced
16	TUFM	Combined oxidative phosphorylation deficiency 4	< 1 in 500	Reduced
6	TULP1	Retinitis pigmentosa 14; Leber congenital amaurosis 15	1 in 1285	1 in 64250
8	TUSC3	Mental retardation, autosomal recessive, type 7	< 1 in 500	Reduced
2	TWIST2	Focal facial dermal dysplasia, type 3 (Setleis type)	< 1 in 500	Reduced
10	TWNK	Mitochondrial DNA depletion syndrome, type 7 (hepatocerebral type); Perrault syndrome type 5	< 1 in 500	Reduced
18	TXNL4A	Burn-McKeown syndrome	< 1 in 500	Reduced
19	TYK2	Immunodeficiency, type 35	< 1 in 500	Reduced
22	TYMP	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	1 in 425	1 in 10625
11	TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	1 in 92	1 in 1840
19	TYROBP	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy, type 1 (Nasu-Hakola disease)	< 1 in 500	Reduced
9	TYRP1	Albinism, oculocutaneous, type 3	< 1 in 500	<1 in 1400
3	UBA5	Epileptic encephalopathy, early infantile, 44	< 1 in 500	Reduced
1	UBE2T	Fanconi anemia, complementation group T	< 1 in 500	Reduced
15	UBE3A	Angelman syndrome	N/A due to imprinting mechanis	Reduced
12	UBE3B	Kaufman oculocerebrofacial syndrome	< 1 in 500	Reduced
15	UBR1	Johanson-Blizzard syndrome	< 1 in 500	Reduced
4	UCHL1	Spastic paraplegia, type 79, autosomal recessive	< 1 in 500	Reduced
13	UFM1	Leukodystrophy, hypomyelinating, type 14	< 1 in 500	Reduced
2	UGT1A1	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	1 in 500	1 in 5496
3	UMPS	Orotic aciduria	< 1 in 500	Reduced
17	UNC13D	Hemophagocytic lymphohistiocytosis, familial, type 3	1 in 108	1 in 202
2	UNC80	Hypotonia, infantile, with psychomotor retardation and characteristic facies, type 2	< 1 in 500	Reduced
12	UNG	Immunodeficiency with hyper IgM, type 5	< 1 in 500	Reduced
22	UPB1	Beta-ureidopropionase deficiency	< 1 in 500	Reduced
8	UQCRB	Mitochondrial complex III deficiency, nuclear, type 3	< 1 in 500	Reduced
16	UQCRC2	Mitochondrial complex III deficiency, nuclear type 5	< 1 in 500	Reduced
5	UQCRQ	Mitochondrial complex III deficiency, nuclear, type 4	< 1 in 500	Reduced
1	UROD	Porphyria cutanea tarda	< 1 in 500	Reduced
10	UROS	Porphyria, congenital erythropoietic	< 1 in 500	Reduced
16	USB1	Poikiloderma with neutropenia	< 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 60	1 in 600
22	USP18	Pseudo-TORCH syndrome 2	< 1 in 500	Reduced
4	UVSSA	UV-sensitive syndrome, type 3	< 1 in 500	Reduced
16	VAC14	Striatonigral degeneration, childhood-onset	< 1 in 500	Reduced
6	VARS1	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy	< 1 in 500	Reduced
6	VARS2	Combined oxidative phosphorylation deficiency 20	< 1 in 500	Reduced
12	VDR	Rickets, vitamin D-resistant, type 2A	< 1 in 500	Reduced
14	VIPAS39	Arthrogryposis, renal dysfunction and cholestasis, type 2	< 1 in 500	Reduced
16	VKORC1	Vitamin K-dependent clotting factors, combined deficiency of, type 2	< 1 in 500	Reduced
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
8	VPS13B	Cohen syndrome	1 in 224	1 in 747
15	VPS13C	Parkinson disease 23, autosomal recessive, early onset	< 1 in 500	Reduced
15	VPS33B	Arthrogryposis, renal dysfunction and cholestasis, type 1	< 1 in 500	1 in 25000
8	VPS37A	Spastic paraplegia, type 53, autosomal recessive	< 1 in 500	Reduced
1	VPS45	Neutropenia, severe congenital, type 5	< 1 in 500	Reduced
17	VPS53	Pontocerebellar hypoplasia, type 2E	1 in 1506	Reduced
14	VRK1	Pontocerebellar hypoplasia, type 1A	< 1 in 500	Reduced
14	VSX2	Microphthalmia with coloboma 3; Isolated microphthalmia 2	1 in 1337	1 in 8913
12	VWF	von Willibrand disease, type 3	1 in 500	1 in 16666
1	WARS2	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures	< 1 in 500	Reduced
12	WASHC4	?Mental retardation, autosomal recessive, type 43	< 1 in 500	Reduced
8	WASHC5	Ritscher-Schinzel syndrome, type 1	< 1 in 500	Reduced
4	WDR19	Nephronophthisis, type 13; Senior-Loken syndrome, type 8	< 1 in 500	Reduced
2	WDR35	Cranioectodermal dysplasia 2	< 1 in 500	Reduced
17	WDR45B	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	< 1 in 500	Reduced
19	WDR62	Microcephaly, type 2, primary, autosomal recessive, with or without cortical malformations	< 1 in 500	Reduced
15	WDR72	Amelogenesis imperfecta, type 2A3 (hypomaturation type)	< 1 in 500	Reduced
15	WDR73	Galloway-Mowat syndrome 1	< 1 in 500	Reduced
17	WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome, type 2	< 1 in 500	Reduced
7	WEE2	Oocyte maturation defect 5	< 1 in 500	Reduced
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 1000	Reduced
2	WIPF1	?Wiskott-Aldrich syndrome 2	< 1 in 500	Reduced
12	WNK1	Neuropathy, hereditary sensory and autonomic, type 2	< 1 in 500	Reduced
12	WNT1	Osteogenesis imperfecta, type XV	< 1 in 500	Reduced
2	WNT10A	WNT10A-related conditions	1 in 238	1 in 2975
12	WNT10B	Split-hand/foot malformation, type 6	< 1 in 500	Reduced
17	WNT3	?Tetra-amelia syndrome	< 1 in 500	Reduced
3	WNT7A	Fuhrmann syndrome	< 1 in 500	Reduced
17	WRAP53	Dyskeratosis congenita, autosomal recessive, type 3	< 1 in 500	Reduced
8	WRN	Werner syndrome	1 in 474	1 in 681
16	WWOX	Epileptic encephalopathy, early infantile, type 28; Spinocerebellar ataxia, autosomal recessive, type 12	< 1 in 500	1 in 16666
2	XDH	Xanthinuria, type 1	< 1 in 500	Reduced
9	XPA	Xeroderma pigmentosum, group A	< 1 in 500	Reduced
3	XPC	Xeroderma pigmentosum, group C	< 1 in 500	Reduced
22	XPNPEP3	Nephronophthisis-like nephropathy, type 1	< 1 in 500	Reduced
5	XRCC4	Short stature, microcephaly, and endocrine dysfunction	< 1 in 500	Reduced
16	XYLT1	Desbuquois dysplasia, type 2	< 1 in 500	Reduced
17	XYLT2	Spondyloocular syndrome	< 1 in 500	Reduced
12	YARS2	Myopathy, lactic acidosis, and sideroblastic anemia, type 2	< 1 in 500	Reduced
1	YY1AP1	Grange syndrome	< 1 in 500	Reduced
2	ZAP70	Autoimmune disease, multisystem, infantile-onset, type 2; Immunodeficiency, type 48	< 1 in 500	Reduced
11	ZBTB16	Skeletal defects, genital hypoplasia, and mental retardation	< 1 in 500	Reduced
6	ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 2	< 1 in 500	Reduced
14	ZC3H14	Mental retardation, autosomal recessive, type 56	< 1 in 500	Reduced
14	ZFYVE26	Spastic paraplegia, type 15, autosomal recessive	< 1 in 500	Reduced
1	ZMPSTE24	Mandibuloacral dysplasia with, type B lipodystrophy	< 1 in 500	Reduced
3	ZMYND10	Ciliary dyskinesia, primary, type 22	< 1 in 500	Reduced
11	ZNF408	Retinitis pigmentosa, type 72	< 1 in 500	Reduced
16	ZNF423	Joubert syndrome, type 19	< 1 in 500	Reduced
16	ZNF469	Brittle cornea syndrome, type 1	< 1 in 500	Reduced
17	ZNHIT3	PEHO syndrome	< 1 in 500	Reduced
11	ZP1	Oocyte maturation defect, type 1	< 1 in 500	Reduced

N/A: no data prevalence unknown