CGT Exome v5.4.5

Patient Information		Sample Information			Clinic Information	
Unique pat id.:	0252184	**Sample type:**	Blood		**Clinic:**	WeFIV
Patient name:		**Date of draw:**	21/11/2024		**Doctor:**	CAROLINA BOUTEILLER
Patient DOB:		**Date of receipt:**	26/11/2024			
Ethnic group:	Caucasian	**Report date/time:**	26/12/2024	7:43		
Indication:	No family history					

TEST RESULTS

POSITIVE

The individual is carrier of:

Alpha-1 antitrypsin deficiency

Gene :	SERPINA1	**Allele:**	Het
DNA Change:	NM_000295.5:c.187C>T	**Inheritance:**	AR
Protein change:	p.Arg63Cys	**OMIM phenotype:**	613490
Variant classification:	Pathogenic / Likely Pathogenic		

Bardet-Biedl syndrome, type 5

Gene :	BBS5	**Allele:**	Het
DNA Change:	NM_152384.3:c.54dupC	**Inheritance:**	AR
Protein change:	p.Ala19fs	**OMIM phenotype:**	615983
Variant classification:	Pathogenic		

Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1

Gene :	SCO2	**Allele:**	Het
DNA Change:	NM_001169109.2:c.121_124delCAGG	**Inheritance:**	AR
Protein change:	p.Gln41fs	**OMIM phenotype:**	604377
Variant classification:	Likely Pathogenic		

Ciliary dyskinesia, primary, type 15

Gene :	CCDC40	**Allele:**	Het
DNA Change:	NM_001243342.2:c.3091T>C	**Inheritance:**	AR
Protein change:	p.Ter1031Argext*?	**OMIM phenotype:**	613808
Variant classification:	Likely Pathogenic		

Ciliary dyskinesia, primary, type 3, with or without situs inversus

Gene :	DNAH5	**Allele:**	Het
DNA Change:	NM_001369.3:c.10815delT	**Inheritance:**	AR
Protein change:	p.Pro3606fs	**OMIM phenotype:**	608644
Variant classification:	Pathogenic		

Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset

Gene :	SLC25A13	**Allele:**	Het
DNA Change:	NM_001160210.2:c.469G>C	**Inheritance:**	AR
Protein change:	p.Glu157Gln	**OMIM phenotype:**	605814; 603471
Variant classification:	Likely Pathogenic		

Deafness, autosomal recessive, type 93

Gene :	CABP2	**Allele:**	Het
DNA Change:	NM_001318496.2:c.304G>A	**Inheritance:**	AR
Protein change:	p.Gly102Ser	**OMIM phenotype:**	614899
Variant classification:	Likely Pathogenic		

Nephrotic syndrome, type 2

Gene :	NPHS2	**Allele:**	Het
DNA Change:	NM_014625.4:c.686G>A	**Inheritance:**	AR
Protein change:	p.Arg229Gln	**OMIM phenotype:**	600995
Variant classification:	Likely Pathogenic		

Prothrombin deficiency

Gene :	F2	**Allele:**	Het
DNA Change:	NM_000506.5:c.1292G>A	**Inheritance:**	AR
Protein change:	p.Arg431His	**OMIM phenotype:**	613679
Variant classification:	Likely Pathogenic		

Spinocerebellar ataxia, autosomal recessive, type 1

Gene :	SETX	**Allele:**	Het
DNA Change:	NM_001351528.2:c.1669C>T	**Inheritance:**	AR
Protein change:	p.Arg557*	**OMIM phenotype:**	606002
Variant classification:	Pathogenic		

Thrombophilia due to antithrombin III deficiency

Gene :	SERPINC1	**Allele:**	Het
DNA Change:	NM_001386302.1:c.1369G>T	**Inheritance:**	AR
Protein change:	p.Ala457Ser	**OMIM phenotype:**	613118*
Variant classification:	Likely Pathogenic		

Usher syndrome, type 1B; Deafness, autosomal recessive, type 2

Gene :	MYO7A	**Allele:**	Het
DNA Change:	NM_000260.4:c.6337G>C	**Inheritance:**	AR
Protein change:	p.Ala2113Pro	**OMIM phenotype:**	276900; 600060
Variant classification:	Likely Pathogenic		

INTERPRETATION OF TEST RESULTS

Typically, a positive result does not have direct clinical consequences for the carrier individual. There is another normal gene copy for all positive autosomal recessive (AR) genes indicated in the table which provides normal biological information. The likelihood of transmission of the variant(s) to offspring is 50%, independent for each variant. If the partner, or gamete donor, screens negative for the pathogenic or likely pathogenic variants in the gene(s) included in the table for this patient, the reproductive risk would be reduced. Please note that family members may also carry the variant(s) reported here, and this information may be significant for them and their offspring.
If a patient and partner, or gamete donor, are both carriers of variants in the same gene associated with AR inheritance, there is a 25% chance that any child they have together would be affected. If a female patient is a carrier for an X-linked condition, there is a 50% chance that each of the reproductive couple's children would also be a carrier. Males would typically express symptoms of the condition, and females are typically unaffected or may display milder symptoms.
For genes with a negative test result, the risk of having children affected by the associated disorders decreases significantly compared to the general population. This also the case for a negative personal result when a reproductive partner or a gamete donor is a carrier for a pathogenic or likely pathogenic variant in one or more of the tested genes. However, due to test limitations associated with any genetic test, this low risk is not zero (see limitations section and informed consent form).

TEST DESCRIPTION

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

COMMENTS

Language changed per clinic request.

TEST METHODOLOGY

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

TEST LIMITATIONS

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

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

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

LEGAL/QUALITY

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: #99#D2146167.

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
	
Camila Ayala Lira da Cruz CRBIO 113163 Bióloga	**Arantxa Hervas PhD** 3025-CV Biotechnologist

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

Alpha-1 antitrypsin deficiency

What is Alpha-1 antitrypsin deficiency?

Alpha-1-antitrypsin deficiency (AATD) is an autosomal recessive disorder caused by pathogenic variants in the SERPINA1 gene. The clinical features can vary but typically involve lung disease and liver disease.

It is important to note that many individuals with pathogenic variants in the SERPINA1 gene will have mild features or remain asymptomatic. There are two common gene variants, often referred to as PI*Z (Z allele) and PI*S (S allele). Other variants in the SERPINA1 gene, including null alleles, are rarer but have the potential to be more severe. Individuals who are homozygous PI*ZZ are at the highest risk for lung and liver disease. In contrast, the PI*SS genotype is not associated with clinical disease. Individuals with the PI*SZ genotype may have an increased risk for lung and liver disease but many individuals with this genotype do not show symptoms.

The earliest symptoms of lung disease begin in early to mid-adulthood and may progress to chronic obstructive pulmonary disease (COPD), which includes emphysema. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema developing at an earlier age. In non-smokers, emphysema may be diagnosed in the sixth or seventh decade of life.

A less common manifestation of AATD is liver disease, which is seen in approximately 15% of individuals. Liver disease may occur at any age and can progress to cirrhosis and liver failure. Some infants with AATD may present with jaundice (yellowing of the skin). The risk for an individual with the PI*ZZ genotype to develop severe liver disease in childhood is generally low (~2%), based on available evidence. If liver disease develops, regular monitoring of liver function and treatment of symptoms is recommended. Some individuals may require a liver transplant if significant damage occurs.

In rare cases, people with alpha-1 antitrypsin deficiency develop a skin condition called panniculitis, which is characterized by hardened skin with painful lumps or patches. Panniculitis varies in severity and can occur at any age.

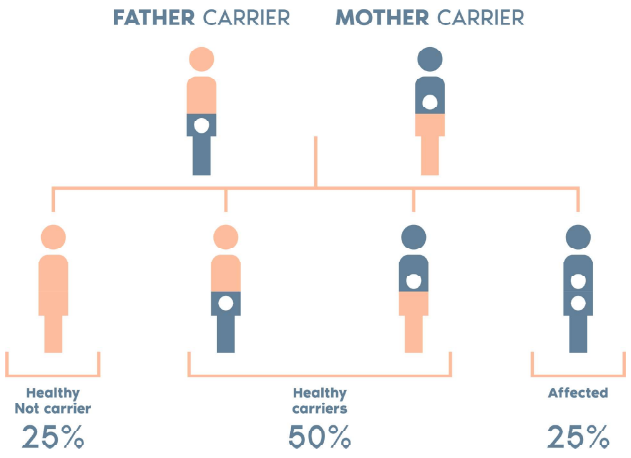
Carriers of AATD are generally healthy. However, carriers of PI*Z who smoke or have other environmental exposures appear to be at an increased risk of developing lung disease.

What is the next step if I am a carrier of Alpha-1 antitrypsin deficiency?

If you are a carrier of Alpha-1 antitrypsin deficiency it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

If your partner tests negative for Alpha-1 antitrypsin deficiency, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.



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

What if both parents are carriers of Alpha-1 antitrypsin deficiency?

When both parents are carriers of Alpha-1 antitrypsin deficiency, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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

Bardet-Biedl syndrome, type 5

What is Bardet-Biedl syndrome, type 5?

Bardet-Biedl syndrome 5 (BBS5) is a ciliopathy associated with severe and early-onset retinal dystrophy, postaxial polydactyly, obesity, renal dysfunction, hypogonadism, and learning difficulties (Scheidecker et al., 2015). Patients described by Young et al. (1999) and Moore et al. (2005) with mutations in the BBS5 gene did not have polydactyly. The contribution of BBS5 mutations to all cases of BBS has been estimated at 2% (Li et al., 2004) and 0.40% (Zaghloul and Katsanis, 2009).

What is the next step if I am a carrier of Bardet-Biedl syndrome, type 5?

If you are a carrier of Bardet-Biedl syndrome, type 5 it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

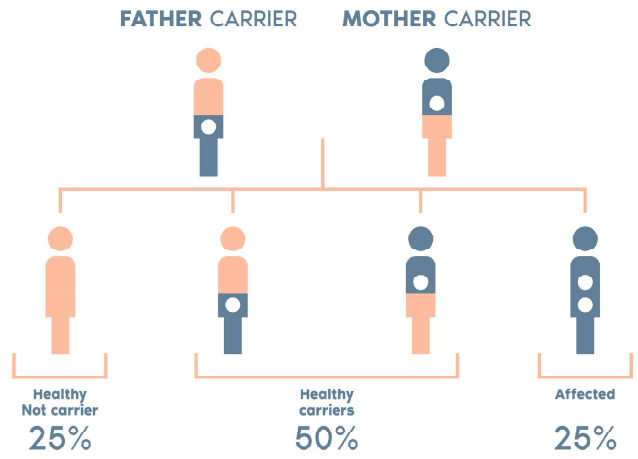
If your partner tests negative for Bardet-Biedl syndrome, type 5, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Bardet-Biedl syndrome, type 5?

When both parents are carriers of Bardet-Biedl syndrome, type 5, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1

What is Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1?

Cardioencephalomyopathy due to cytochrome c oxidase deficiency is an autosomal recessive mitochondrial disorder characterized by onset of cardiomyopathy either in utero or in the first days of life. Most patients also show neurologic abnormalities, such as abnormal breathing pattern, nystagmus, and gyral abnormalities, consistent with encephalopathy. The disorder is usually fatal in early infancy (Papadopoulou et al., 1999).

What is the next step if I am a carrier of Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1?

If you are a carrier of Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1 it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

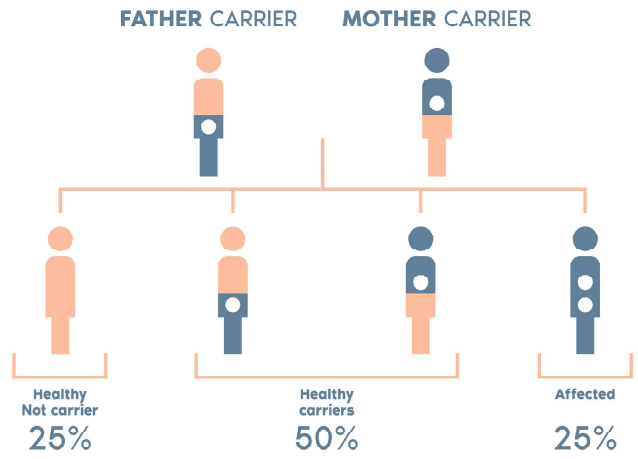
If your partner tests negative for Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1?

When both parents are carriers of Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

Ciliary dyskinesia, primary, type 15

What is Ciliary dyskinesia, primary, type 15?

Ciliary dyskinesia, primary, type 15 (CILD15) is an autosomal recessive disorder characterized by recurrent respiratory infections associated with defects in ciliary inner dynein arms and axonemal disorganization (Becker-Heck et al., 2011).

What is the next step if I am a carrier of Ciliary dyskinesia, primary, type 15?

If you are a carrier of Ciliary dyskinesia, primary, type 15 it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

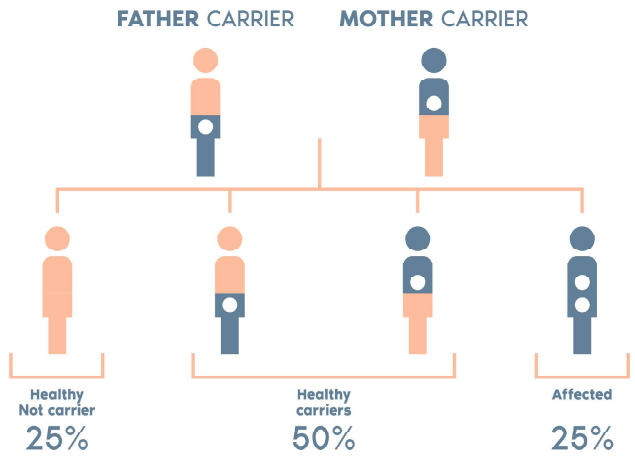
If your partner tests negative for Ciliary dyskinesia, primary, type 15, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Ciliary dyskinesia, primary, type 15?

When both parents are carriers of Ciliary dyskinesia, primary, type 15, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

Ciliary dyskinesia, primary, type 3, with or without situs inversus

What is Ciliary dyskinesia, primary, type 3, with or without situs inversus?

Ciliary dyskinesia, primary, type 3 (CILD3) is an autosomal recessive disorder resulting from loss of normal ciliary function. Kartagener (pronounced KART-agayner) syndrome is characterized by the combination of primary ciliary dyskinesia and situs inversus, and occurs in approximately half of patients with ciliary dyskinesia. Since normal nodal ciliary movement in the embryo is required for normal visceral asymmetry, absence of normal ciliary movement results in a lack of definitive patterning; thus, random chance alone appears to determine whether the viscera take up the normal or reversed left-right position during embryogenesis. This explains why approximately 50% of patients, even within the same family, have situs inversus (Afzelius, 1976; El Zein et al., 2003).

What is the next step if I am a carrier of Ciliary dyskinesia, primary, type 3, with or without situs inversus?

If you are a carrier of Ciliary dyskinesia, primary, type 3, with or without situs inversus it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

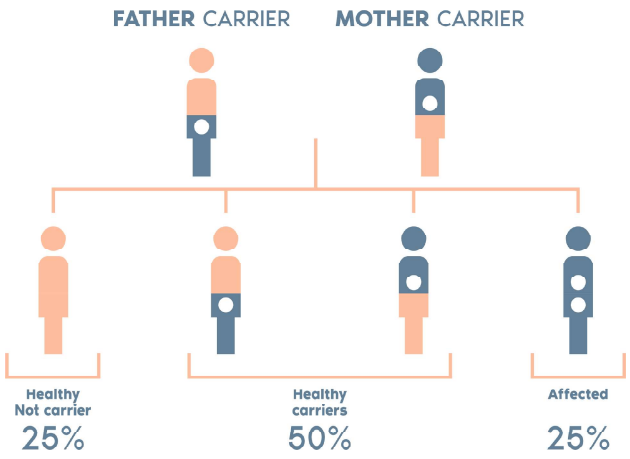
If your partner tests negative for Ciliary dyskinesia, primary, type 3, with or without situs inversus, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Ciliary dyskinesia, primary, type 3, with or without situs inversus?

When both parents are carriers of Ciliary dyskinesia, primary, type 3, with or without situs inversus, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset

What is Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset?

Neonatal-onset type II citrullinemia, also known as neonatal intrahepatic cholestasis, is caused by homozygous or compound heterozygous mutation in the SLC25A13 gene (603859). Adult-onset type II citrullinemia (603471) is caused by mutation in the same gene.
Neonatal intrahepatic cholestasis caused by citrin deficiency (NICCD) is an autosomal recessive metabolic disorder characterized by poor growth, intrahepatic cholestasis, and increased serum citrulline. Most patients show spontaneous improvement by 1 year of age. However, some patients may have a progressive course with continued failure to thrive and dyslipidemia caused by citrin deficiency (FTTDCD), and some may develop chronic or fatal liver disease (summary by Song et al., 2011).
Adult-onset type II citrullinemia is an autosomal recessive metabolic disorder characterized clinically by the sudden onset of various neuropsychologic symptoms such as disorientation, abnormal behavior, convulsions, and coma due to hyperammonemia. In some cases, rapid progression can lead to brain edema and death if liver transplantation is not possible. Some patients may present with nonalcoholic hepatic steatosis or may develop hepatic fibrosis or hepatocellular carcinoma. Patients with this disorder have a natural aversion to carbohydrates and favor protein, which is in contrast to protein aversion usually observed in patients with urea cycle defects (summary by Komatsu et al., 2008).

What is the next step if I am a carrier of Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset?

If you are a carrier of Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

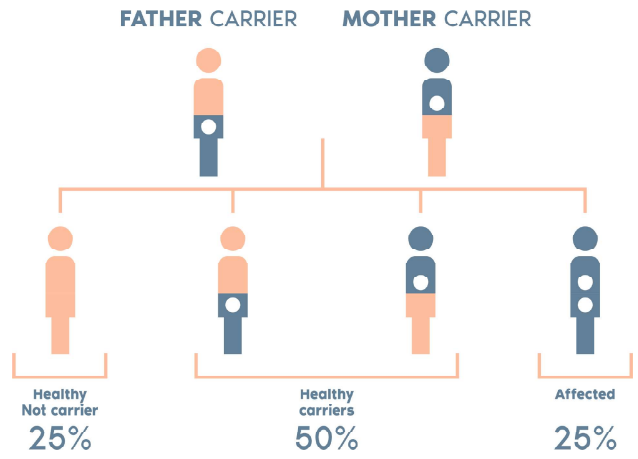
If your partner tests negative for Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset?

When both parents are carriers of Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

Deafness, autosomal recessive, type 93

What is Deafness, autosomal recessive, type 93?

Nonsyndromic hearing loss is a partial or total loss of hearing that is not associated with other signs and symptoms. Nonsyndromic hearing loss can be classified by the condition's pattern of inheritance: autosomal dominant (DFNA), autosomal recessive (DFNB), X-linked (DFNX), or mitochondrial (which does not have a special designation). DFNA, DFNB, and DFNX subtypes are numbered in the order in which they were first described. The characteristics vary among the different types. Hearing loss can affect one ear (unilateral) or both ears (bilateral). Degrees of hearing loss range from mild (difficulty understanding soft speech) to profound (inability to hear even very loud noises). The term "deafness" is often used to describe severe-to-profound hearing loss. Hearing loss can be stable, or it may be progressive, becoming more severe as a person gets older. Particular types of nonsyndromic hearing loss show distinctive patterns of hearing loss. Most forms of nonsyndromic hearing loss are described as sensorineural, which means they are associated with a permanent loss of hearing caused by damage to structures in the inner ear.

What is the next step if I am a carrier of Deafness, autosomal recessive, type 93?

If you are a carrier of Deafness, autosomal recessive, type 93 it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

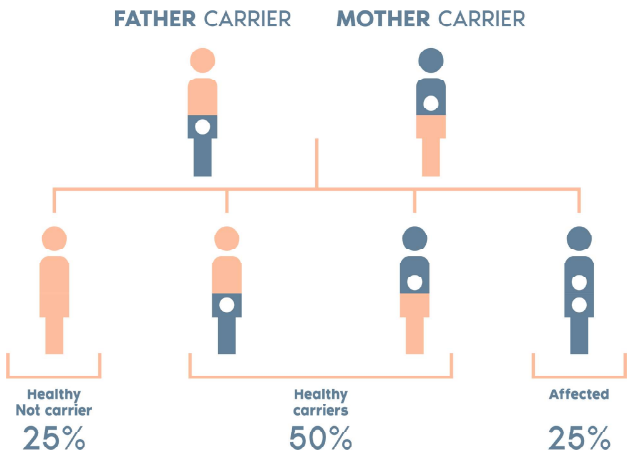
If your partner tests negative for Deafness, autosomal recessive, type 93, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Deafness, autosomal recessive, type 93?

When both parents are carriers of Deafness, autosomal recessive, type 93, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

Nephrotic syndrome, type 2

What is Nephrotic syndrome, type 2?

Nephrotic syndrome, type 2 (NPHS2) is an autosomal recessive disorder characterized clinically by childhood onset of proteinuria, hypoalbuminemia, hyperlipidemia, and edema. Kidney biopsies show nonspecific histologic changes such as minimal change, focal segmental glomerulosclerosis (FSGS), and diffuse mesangial proliferation. The disorder is resistant to steroid treatment and progresses to end-stage renal failure in the first or second decades (Fuchshuber et al., 1996). Some patients show later onset of the disorder (Tsukaguchi et al., 2002).

What is the next step if I am a carrier of Nephrotic syndrome, type 2?

If you are a carrier of Nephrotic syndrome, type 2 it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

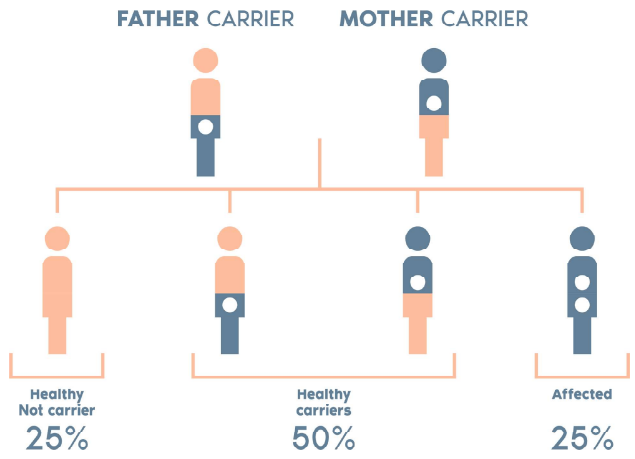
If your partner tests negative for Nephrotic syndrome, type 2, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Nephrotic syndrome, type 2?

When both parents are carriers of Nephrotic syndrome, type 2, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

Prothrombin deficiency

What is Prothrombin deficiency?

Prothrombin deficiency is an extremely rare autosomal recessive bleeding disorder characterized by low levels of circulating prothrombin; it affects about 1 in 2,000,000 individuals. There are 2 main types: type 1 deficiency, known as true prothrombin deficiency or 'hypoprothrombinemia,' is defined as plasma levels of prothrombin being less than 10% of normal with a concomitant decrease in activity. These patients have severe bleeding from birth, including umbilical cord hemorrhage, hematomas, ecchymoses, hematuria, mucosal bleeding, hemarthroses, intracranial bleeding, gastrointestinal bleeding, and menorrhagia. Type 2 deficiency, known as 'dysprothrombinemia,' is characterized by normal or low-normal synthesis of a dysfunctional protein. Bleeding symptoms are more variable, depending on the amount of residual functional activity. Variant prothrombin gene alleles can result in 'hypoprothrombinemia' or 'dysprothrombinemia,' and individuals who are compound heterozygous for these 2 types of alleles have variable manifestations. Heterozygous mutation carriers, who have plasma levels between 40 and 60% of normal, are usually asymptomatic, but can show bleeding after tooth extraction or surgical procedures (Lancellotti and De Cristofaro, 2009).

What is the next step if I am a carrier of Prothrombin deficiency?

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

What if my partner isn't a carrier?

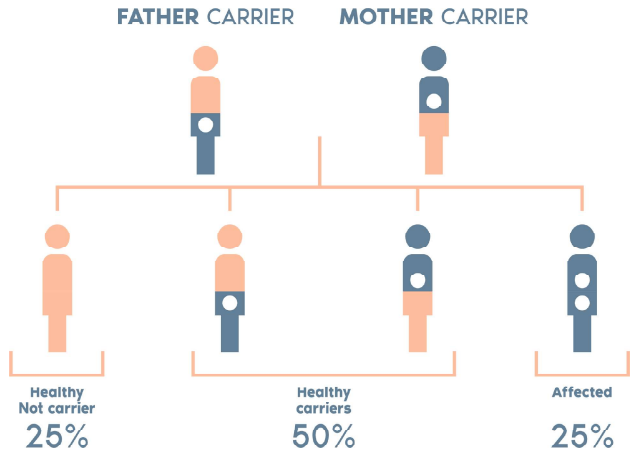
If your partner tests negative for Prothrombin deficiency, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk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

What if both parents are carriers of Prothrombin deficiency?

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

What if I am going to use gamete donation?

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



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

Spinocerebellar ataxia, autosomal recessive, type 1

What is Spinocerebellar ataxia, autosomal recessive, type 1?

Spinocerebellar ataxia with axonal neuropathy type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the SETX gene located on chromosomal region 9q34.13. The age of onset is infantile. This disease is characterized by progressive cerebellar ataxia, axonal sensorimotor neuropathy with oculomotor apraxia, fixation instability, extrapyramidal features and an elevated serum alpha-fetoprotein level. The prevalence is 4:100,000-8:100,000.

What is the next step if I am a carrier of Spinocerebellar ataxia, autosomal recessive, type 1?

If you are a carrier of Spinocerebellar ataxia, autosomal recessive, type 1 it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

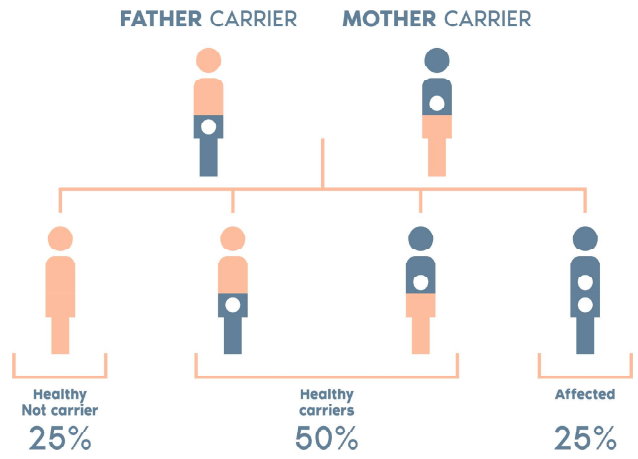
If your partner tests negative for Spinocerebellar ataxia, autosomal recessive, type 1, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Spinocerebellar ataxia, autosomal recessive, type 1?

When both parents are carriers of Spinocerebellar ataxia, autosomal recessive, type 1, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

Thrombophilia due to antithrombin III deficiency

What is Thrombophilia due to antithrombin III deficiency?

Deficiency of antithrombin III is a major risk factor for venous thromboembolic disease. Two categories of AT-III deficiency have been defined on the basis of AT-III antigen levels in the plasma of affected individuals. The majority of AT-III deficiency families belong in the type I (classic) deficiency group and have a quantitatively abnormal phenotype in which antigen and heparin cofactor levels are both reduced to about 50% of normal. The second category of AT-III deficiency has been termed type II (functional) deficiency. Affected individuals from these kindreds produce dysfunctional AT-III molecules; they have reduced heparin cofactor activity levels (about 50% of normal) but levels of AT-III antigen are often normal or nearly normal (Bock and Prochownik, 1987).The 2 categories of antithrombmin III deficiency have been classified further. Type I (low functional and immunologic antithrombin) has been subdivided into subtype Ia (reduced levels of normal antithrombin), and type Ib (reduced levels of antithrombin and the presence of low levels of a variant). Type II (low functional but normal immunologic antithrombin) has been subdivided into subtype IIa (functional abnormalities affecting both the reactive site and the heparin-binding site of AT3); subtype IIb (functional abnormalities limited to the reactive site); and subtype IIc (functional abnormalities limited to the heparin-binding site) (Lane et al., 1992).

What is the next step if I am a carrier of Thrombophilia due to antithrombin III deficiency?

If you are a carrier of Thrombophilia due to antithrombin III deficiency it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

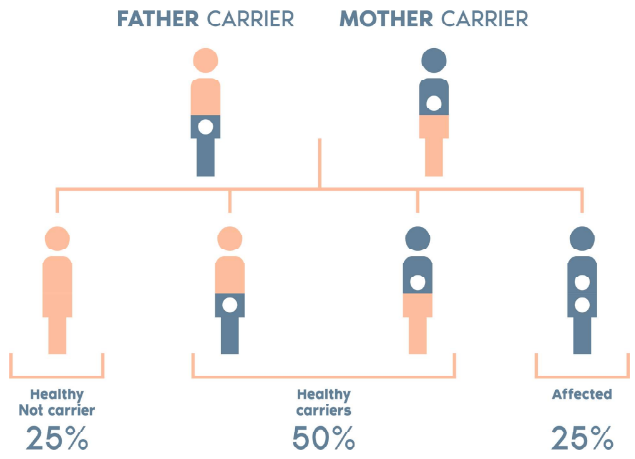
If your partner tests negative for Thrombophilia due to antithrombin III deficiency, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants.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

What if both parents are carriers of Thrombophilia due to antithrombin III deficiency?

When both parents are carriers of Thrombophilia due to antithrombin III deficiency, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

Usher syndrome, type 1B; Deafness, autosomal recessive, type 2

What is Usher syndrome, type 1B; Deafness, autosomal recessive, type 2?

Usher syndrome type 1B follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the MYO7A gene located on chromosomal region 11q13.5. The age of onset is infantile. This disease is characterized by congenital, bilateral, profound sensorineural hearing loss, vestibular areflexia, and adolescent-onset retinitis pigmentosa. The prevalence is 1:100,000-9:100,000. Mutation in the MYO7A gene can also cause deafness autosomal recesive, type 2 which involves changes in both the inner ear and the middle ear. This combination is called mixed hearing loss.

What is the next step if I am a carrier of Usher syndrome, type 1B; Deafness, autosomal recessive, type 2?

If you are a carrier of Usher syndrome, type 1B; Deafness, autosomal recessive, type 2 it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

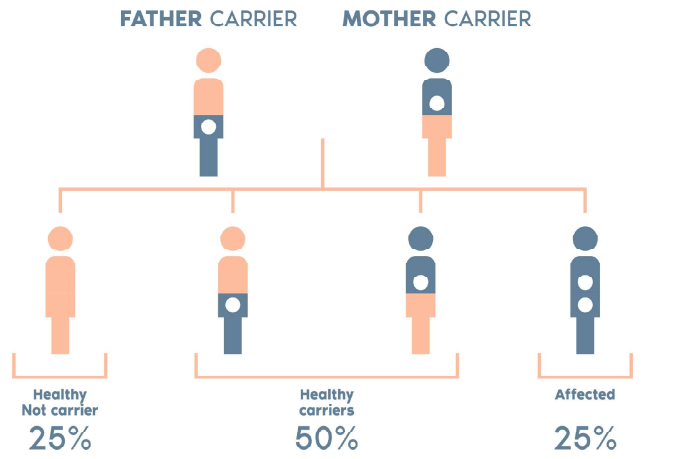
If your partner tests negative for Usher syndrome, type 1B; Deafness, autosomal recessive, type 2, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Usher syndrome, type 1B; Deafness, autosomal recessive, type 2?

When both parents are carriers of Usher syndrome, type 1B; Deafness, autosomal recessive, type 2, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

LIST OF ANALYZED GENES

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, 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SLC26A2, CLN6, SLC12A5, SAMHD1, DUOX2, TPO, TKT, ST14, GAA, FTCD, MYO3A, PRODH, ALDH4A1, COG5, POMGNT1, ALMS1, SERPING1, HEXB, PHGDH, ATP7B, IRAK4, ACADS, SUOX, GALC, LYST, TYR, NDUFAF1, UROS, ALG9, LDLR, GALE, LCAT, COG1, COG4, COG6, COG7, COQ8A, MPO, GALT, ACADM, TRPM6, SUFU, IVD, OTOA, IMPG2, USP18, SLC39A4, FKBP10, ZMYND10, SLC22A12, NPHP1, SPART, PLCB1, FANCA, DTNBP1, SEC24D, TDP1, ALOXE3, STUB1, CARD11, CARD9, ORC6, NPHP4, SMOC2, AP4E1, AP4B1, AP3D1, EVC2, SEMA4A, MYO18B, CABP2, TRAF3IP1, IFT172, MC2R, NME8, POMT1, PDSS1, GTPBP2, ERAL1, POMT2, FKTN, SBDS, CDAN1, HGD, MMAA, ADAMTS17, ADAMTS18, HPS5, PIBF1, LRPPRC, MSMO1, MMAB, ATM, BBS7, KCNV2, SMPD1, NUP107, NPC1, XDH, OSTM1, CTH, GNS, SECISBP2, USH1G, SBF2, KLHL41, TJP2, ITGA2B, ABCA12, CNNM4, ACAT1, VPS13B, FRAS1, GBE1, GNPTAB, BEST1, YY1AP1, INPP5K, NNT, DSG4, FERMT1, FERMT3, ALG2, WHRN, SUMF1, CEP57, BBS9, SCYL1, GORAB, TPP1, NPHP3, SIL1, NBAS, ANTXR2, UBE3B, ETFA, APOC2, NFU1, ALG8, MEFV, FANCL, WDR19, CACNA2D4, DHDDS, 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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, HFE, SCARF2, ALG11, SOBP, GZF1, BPGM, SPATA5, 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 50,000	1 in 333333
X	CD40LG	Hyper-IgM syndrome, type 1 (immunodeficiency, X-linked, with hyper-IgM, type 1)	< 1 in 100 000	Reduced
X	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 1374	1 in 27480
X	EDA	Ectodermal dysplasia, type 1, hypohidrotic, X-linked	1 in 2500	1 in 16667
X	EMD	Emery-Dreifuss muscular dystrophy, type 1, X-linked	< 1 in 100 000	Reduced
X	F8	Hemophilia A	1 in 1250	1 in 31250
X	F9	Hemophilia B	1 in 6250	1 in 62500
X	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 50,000	<1 in 1,000,000
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 556,112	<1 in 1,000,000
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	Mental retardation, 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 8,266
16	AARS1	Epileptic encephalopathy, early infantile, type 29	N/A	N/A
6	AARS2	Combined oxidative phosphorylation deficiency 8; Leukoencephalopathy, progressive, with ovarian failure	N/A	N/A
7	AASS	Hyperlysinemia, type 1 and type 2	N/A	N/A
16	ABAT	GABA-transaminase deficiency	< 1 in 500	Reduced
9	ABCA1	Tangier disease	N/A	N/A
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 7,143
1	ABCA4	Stargardt disease 1; Retinitis pigmentosa 19; Cone-rod dystrophy 3	1 in 62	1 in 119
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	N/A	N/A
10	ABCC2	Dubin-Johnson syndrome	N/A	N/A
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 49,501
2	ABCG5	Sitosterolemia 2	N/A	N/A
2	ABCG8	Sitosterolemia 1	N/A	N/A
20	ABHD12	PHARC syndrome (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa and cataract)	N/A	N/A
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 94	1 in 1880
10	ACADSB	Short/branched-chain acyl-CoA dehydrogenase deficiency	1 in 500	1 in 1,125
17	ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	1 in 112	1 in 1120
11	ACAT1	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	1 in 300	1 in 3750
17	ACE	Renal tubular dysgenesis	< 1 in 500	Reduced
22	ACO2	Infantile cerebellar-retinal degeneration	N/A	N/A
17	ACOX1	Peroxisomal acyl-CoA oxidase deficiency	< 1 in 500	Reduced
3	ACOX2	Bile acid synthesis defect, congenital, type 6	N/A	N/A
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	N/A	N/A
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	N/A	N/A
8	ADAM9	Cone-rod dystrophy 9	< 1 in 500	Reduced
19	ADAMTS10	Weill-Marchesani syndrome, type 1, recessive	N/A	N/A
9	ADAMTS13	Thrombotic thrombocytopenic purpura, familial (Schulman-Upshaw syndrome)	N/A	N/A
15	ADAMTS17	Weill-Marchesani syndrome, type 4, recessive	< 1 in 500	Reduced
16	ADAMTS18	Microcornea, myopic chorioretinal atrophy, and telecanthus	N/A	N/A
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	N/A	N/A
1	ADAR	Aicardi-Goutieres syndrome, type 6	N/A	N/A
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 147
10	ADK	Hypermethioninemia due to adenosine kinase deficiency	1 in 500	1 in 1,498
22	ADSL	Adenylosuccinase deficiency	N/A	N/A
14	ADSS1	Myopathy, distal, 5	N/A	N/A
4	AFG2A	Epilepsy, hearing loss, and mental retardation syndrome	< 1 in 500	Reduced
18	AFG3L2	Spastic ataxia, type 5, autosomal recessive	N/A	N/A
4	AFP	Alpha-fetoprotein deficiency	< 1 in 500	Reduced
4	AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)	< 1 in 500	Reduced
2	AGBL5	Retinitis pigmentosa 75	N/A	N/A
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	N/A	N/A
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 571
21	AIRE	Autoimmune polyendocrinopathy syndrome, type 1	1 in 310	1 in 4429
9	AK1	Hemolytic anemia due to adenylate kinase deficiency	N/A	N/A
1	AK2	Reticular dysgenesis	< 1 in 500	Reduced
10	AKR1C2	46,XY disorder of sex development due to testicular 17,20-desmolase deficiency	N/A	N/A
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)	N/A	N/A
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 49,951
6	ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	N/A	N/A
14	ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency	N/A	N/A
5	ALDH7A1	Epilepsy, pyridoxine-dependent	N/A	N/A
16	ALDOA	Glycogen storage disease type 12	< 1 in 500	Reduced
9	ALDOB	Fructose intolerance, hereditary	1 in 80	1 in 400
16	ALG1	Congenital disorder of glycosylation, type 1K	1 in 87	1 in 130
13	ALG11	Congenital disorder of glycosylation, type 1P	< 1 in 500	Reduced
22	ALG12	Congenital disorder of glycosylation, type 1G	N/A	N/A
9	ALG2	Myasthenic syndrome, congenital, type 14, with tubular aggregates	N/A	N/A
3	ALG3	Congenital disorder of glycosylation, type 1D	N/A	N/A
1	ALG6	Congenital disorder of glycosylation, type 1C	1 in 500	1 in 5000
11	ALG8	Congenital disorder of glycosylation, type 1H	N/A	N/A
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	N/A	N/A
17	ALOXE3	Ichthyosis, congenital, autosomal recessive, type 3	N/A	N/A
15	ALPK3	Cardiomyopathy, familial hypertrophic, type 27	N/A	N/A
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	N/A	N/A
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	N/A	N/A
4	AMBN	Amelogenesis imperfecta, type IF	< 1 in 500	Reduced
19	AMH	Persistent Mullerian duct syndrome, type 1	N/A	N/A
12	AMHR2	Persistent Mullerian duct syndrome, type II	N/A	N/A
14	AMN	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)	N/A	N/A
1	AMPD1	Myopathy due to myoadenylate deaminase deficiency	N/A	N/A
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	N/A	N/A
9	ANKS6	Nephronophthisis 16	N/A	N/A
3	ANO10	Spinocerebellar ataxia, autosomal recessive, type 10	1 in 224	1 in 2,236
11	ANO5	Limb-girdle muscular dystrophy, type 12 (LGMD R12)	< 1 in 500	Reduced
2	ANTXR1	GAPO syndrome	N/A	N/A
4	ANTXR2	Hyaline fibromatosis syndrome	N/A	N/A
7	AP1S1	MEDNIK syndrome	< 1 in 500	Reduced
5	AP3B1	Hermansky-Pudlak syndrome, type 2	N/A	N/A
15	AP3B2	Epileptic encephalopathy, early infantile, type 48	< 1 in 500	Reduced
19	AP3D1	Hermansky-Pudlak syndrome, type 10	N/A	N/A
1	AP4B1	Spastic paraplegia, type 47, autosomal recessive	N/A	N/A
15	AP4E1	Spastic paraplegia, type 51, autosomal recessive	< 1 in 500	Reduced
7	AP4M1	Spastic paraplegia, type 50, autosomal recessive	N/A	N/A
14	AP4S1	Spastic paraplegia, type 52, autosomal recessive	1 in 500	1 in 50000
7	AP5Z1	Spastic paraplegia, type 48, autosomal recessive	N/A	N/A
19	APOC2	Hyperlipoproteinemia, type 1B	< 1 in 500	Reduced
19	APOE	Sea-blue histiocyte disease	N/A	N/A
16	APRT	Adenine phosphoribosyltransferase deficiency	N/A	N/A
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	N/A	N/A
19	ARHGEF18	Retinitis pigmentosa 78	N/A	N/A
3	ARL13B	Joubert syndrome type 8	1 in 72	1 in 119
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	N/A	N/A
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	N/A	N/A
8	ASAH1	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy	N/A	N/A
7	ASL	Argininosuccinic aciduria	1 in 133	1 in 665
7	ASNS	Asparagine synthetase deficiency	< 1 in 500	Reduced
17	ASPA	Canavan disease	1 in 416	1 in 13867
8	ASPH	Traboulsi syndrome	N/A	N/A
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	N/A	N/A
16	ATP2A1	Brody myopathy	N/A	N/A
12	ATP6V0A2	Cutis laxa, autosomal recessive, type 2A; Wrinkly skin syndrome	< 1 in 500	Reduced
7	ATP6V0A4	Renal tubular acidosis, distal, autosomal recessive	N/A	N/A
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 1,803
3	ATR	Seckel syndrome, type 1	< 1 in 500	Reduced
9	AUH	3-methylglutaconic aciduria, type 1	< 1 in 500	<1 in 938
19	AURKC	Spermatogenic failure, type 5	N/A	N/A
12	AVIL	Nephrotic syndrome, type 21	N/A	N/A
15	B2M	Immunodeficiency, type 43	< 1 in 500	Reduced
1	B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	N/A	N/A
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	N/A	N/A
13	B3GLCT	Peters-plus syndrome	N/A	N/A
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 50,000
5	B4GALT7	Ehlers-Danlos syndrome, spondylodysplastic, type 1	N/A	N/A
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	N/A	N/A
7	BBS9	Bardet-Biedl syndrome, type 9	N/A	N/A
19	BCAT2	?Hypervalinemia or hyperleucine-isoleucinemia	N/A	N/A
3	BCHE	Butyrylcholinesterase deficiency	1 in 30	<1 in 270
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	N/A	N/A
1	BCL10	?Immunodeficiency, type 37	N/A	N/A
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	N/A	N/A
19	BLOC1S3	Hermansky-Pudlak syndrome, type 8	< 1 in 500	Reduced
15	BLOC1S6	?Hermansky-Pudlak syndrome, type 9	N/A	N/A
4	BLTP1	Alkuraya-Kucinskas syndrome	N/A	N/A
7	BLVRA	Hyperbiliverdinemia	< 1 in 500	Reduced
8	BMP1	Osteogenesis imperfecta, type 13	N/A	N/A
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	N/A	N/A
14	BRF1	Cerebellofaciodental syndrome	N/A	N/A
17	BRIP1	Fanconi anemia, complementation group J	N/A	N/A
11	BSCL2	Congenital generalized lipodystrophy, type 2; Encephalopathy, progressive, with or without lipodystrophy	< 1 in 500	Reduced
1	BSND	Bartter syndrome, type 4A	< 1 in 500	Reduced
3	BTD	Biotinidase deficiency	1 in 120	1 in 1500
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	< 1 in 500	Reduced
12	C1S	C1s deficiency	N/A	N/A
6	C2	C2 deficiency	N/A	N/A
11	C2CD3	Orofaciodigital syndrome, type 14	N/A	N/A
19	C3	Complement component 3 deficiency	N/A	N/A
9	C5	Complement component 5 deficiency	N/A	N/A
5	C6	Complement component 6 deficiency	N/A	N/A
5	C7	Complement component 7 deficiency	N/A	N/A
1	C8B	Complement component 8 deficiency, type 2	N/A	N/A
15	CA12	Hyperchlorhidrosis, isolated	N/A	N/A
8	CA2	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	< 1 in 500	<1 in 1,000
16	CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency	N/A	N/A
8	CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	< 1 in 500	Reduced
11	CABP2	Deafness, autosomal recessive, type 93	N/A	N/A
11	CABP4	Congenital stationary night blindness, type 2B	< 1 in 500	Reduced
3	CACNA1D	Sinoatrial node dysfunction and deafness	N/A	N/A
12	CACNA2D4	Retinal cone dystrophy 4	N/A	N/A
2	CAD	Epileptic encephalopathy, early infantile, 50	N/A	N/A
2	CALCRL	?Lymphatic malformation 8	N/A	N/A
17	CANT1	Desbuquois dysplasia, type 1; Epiphyseal dysplasia, multiple, type 7	N/A	N/A
11	CAPN1	Spastic paraplegia, type 76, autosomal recessive	N/A	N/A
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	N/A	N/A
13	CARS2	Combined oxidative phosphorylation deficiency 27	N/A	N/A
1	CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, type 2	1 in 51	1 in 98
3	CASR	Hyperparathyroidism, neonatal	< 1 in 500	Reduced
5	CAST	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads	N/A	N/A
11	CAT	Acatalasemia	N/A	N/A
11	CATSPER1	Spermatogenic failure, type 7	N/A	N/A
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 2,800
18	CCBE1	Hennekam lymphangiectasia-lymphedema syndrome, type 1	N/A	N/A
17	CCDC103	Ciliary dyskinesia, primary, type 17	N/A	N/A
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	N/A	N/A
17	CCDC40	Ciliary dyskinesia, primary, type 15	N/A	N/A
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 7,143

6	CCN6	Progressive pseudorheumatoid dysplasia	< 1 in 500	Reduced
5	CCNO	Ciliary dyskinesia, primary, type 29	N/A	N/A
16	CD19	Immunodeficiency, common variable, type 3	< 1 in 500	Reduced
1	CD247	?Immunodeficiency, type 25	N/A	N/A
12	CD27	Lymphoproliferative syndrome 2	< 1 in 500	Reduced
6	CD2AP	Glomerulosclerosis, focal segmental, type 3, susceptibility to	N/A	N/A
19	CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect	< 1 in 500	Reduced
7	CD36	Platelet glycoprotein 4 deficiency	N/A	N/A
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)	N/A	N/A
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	N/A	N/A
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	N/A	N/A
10	CDHR1	Cone-rod dystrophy, type 15	< 1 in 500	Reduced
15	CDIN1	Dyserythropoietic anemia, congenital, type Ib	N/A	N/A
16	CDK10	Al Kaissi syndrome	N/A	N/A
9	CDK5RAP2	Primary microcephaly type 3, autosomal recessive	N/A	N/A
6	CDSN	Peeling skin syndrome 1	< 1 in 500	Reduced
16	CDT1	Meier-Gorlin syndrome, type 4	N/A	N/A
14	CEBPE	Specific granule deficiency	< 1 in 500	Reduced
1	CENPF	Stromme syndrome	N/A	N/A
13	CENPJ	Primary microcephaly type 6, autosomal recessive	< 1 in 500	Reduced
1	CEP104	Joubert syndrome 25	N/A	N/A
5	CEP120	Short-rib thoracic dysplasia 13 with or without polydactyly	N/A	N/A
4	CEP135	Microcephaly 8, primary, autosomal recessive	1 in 500	1 in 5000
15	CEP152	Primary microcephaly type 9, autosomal recessive	N/A	N/A
11	CEP164	Nephronophthisis 15	N/A	N/A
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	N/A	N/A
10	CEP55	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly	N/A	N/A
11	CEP57	Mosaic variegated aneuploidy syndrome 2	< 1 in 500	Reduced
9	CEP78	Cone-rod dystrophy and hearing loss	N/A	N/A
12	CEP83	Nephronophthisis 18	N/A	N/A
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	N/A	N/A
18	CFAP53	Heterotaxy, visceral, 6, autosomal recessive	N/A	N/A
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	N/A	N/A
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 121	1 in 134
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	N/A	N/A
17	CHRNB1	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	N/A	N/A
2	CHRND	Myasthenic syndrome, congenital, type 3B, fast-channel; Multiple pterygium syndrome, lethal type	N/A	N/A
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 1,597
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	N/A	N/A
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	N/A	N/A
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	N/A	N/A
16	CLCN7	Osteopetrosis, autosomal recessive type 4	< 1 in 500	Reduced
1	CLCNKA	Bartter syndrome, type 4B, digenic	N/A	N/A
1	CLCNKB	Bartter syndrome, type 3; Bartter syndrome, type 4B, digenic	N/A	N/A
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	N/A	N/A
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 346
13	CLN5	Ceroid lipofuscinosis, neuronal, type 5	1 in 400	1 in 8000
15	CLN6	Ceroid lipofuscinosis, neuronal, type 6	< 1 in 500	Reduced
8	CLN8	Ceroid lipofuscinosis, neuronal, type 8	< 1 in 500	Reduced
11	CLP1	Pontocerebellar hypoplasia, type 10	< 1 in 500	Reduced
11	CLPB	3-methylglutaconic aciduria, type 7, with cataracts, neurologic involvement and neutropenia	N/A	N/A
19	CLPP	Perrault syndrome 3	N/A	N/A
3	CLRN1	Usher syndrome, type 3A	1 in 250	1 in 1667
4	CNGA1	Retinitis pigmentosa type 49	1 in 625	1 in 1,171
2	CNGA3	Achromatopsia, type 2	N/A	N/A
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	N/A	N/A
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	N/A	N/A
17	COG1	Congenital disorder of glycosylation, type IIg	< 1 in 500	Reduced
16	COG4	Congenital disorder of glycosylation, type 2J	N/A	N/A
7	COG5	Congenital disorder of glycosylation, type 2I	N/A	N/A
13	COG6	Congenital disorder of glycosylation, type 2L; Shaheen syndrome	N/A	N/A
16	COG7	Congenital disorder of glycosylation, type 2E	< 1 in 500	Reduced
16	COG8	Congenital disorder of glycosylation, type 2H	N/A	N/A
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	N/A	N/A
10	COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type	< 1 in 500	Reduced
21	COL18A1	Knobloch syndrome, type 1	< 1 in 500	Reduced
7	COL1A2	Ehlers-Danlos syndrome, cardiac valvular type	N/A	N/A
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 2	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])	N/A	N/A
21	COL6A2	Ullrich congenital muscular dystrophy 1; Bethlem myopathy-1; Myosclerosis	N/A	N/A
2	COL6A3	Bethlem myopathy 1; Ullrich congenital muscular dystrophy 1; Dystonia 27	N/A	N/A
3	COL7A1	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	1 in 150	1 in 1000
6	COL9A1	Stickler syndrome, type 4	N/A	N/A
1	COL9A2	?Stickler syndrome, type V	N/A	N/A
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 1,420
4	COQ2	Primary coenzyme Q10 deficiency, type 1	< 1 in 500	Reduced

9	COQ4	Coenzyme Q10 deficiency, primary, type 7	N/A	N/A
14	COQ6	Coenzyme Q10 deficiency, primary, type 6	N/A	N/A
1	COQ8A	Primary coenzyme Q10 deficiency, type 4	< 1 in 500	Reduced
19	COQ8B	Nephrotic syndrome, type 9	N/A	N/A
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 640
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	N/A	N/A
8	CPA6	Febrile seizures, familial, type 11	N/A	N/A
19	CPAMD8	Anterior segment dysgenesis, type 8	N/A	N/A
5	CPLANE1	Joubert syndrome 17	N/A	N/A
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	N/A	N/A
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 316
9	CRB2	Ventriculomegaly with cystic kidney disease	N/A	N/A
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	N/A	N/A
3	CRTAP	Osteogenesis imperfecta, type 7	1 in 1,416	1 in 3,539
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	N/A	N/A
8	CSPP1	Joubert syndrome 21	N/A	N/A
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	N/A	N/A
1	CTH	Cystathioninuria	1 in 80	1 in 7,870
17	CTNS	Nephropathic cystinosis	1 in 200	1 in 400
1	CTPS1	Immunodeficiency, type 24	< 1 in 500	Reduced
20	CTSA	Galactosialidosis	1 in 64	1 in 118
11	CTSC	Haim-Munk syndrome; Papillon-Lefevre syndrome	1 in 500	1 in 2,496
11	CTSD	Ceroid lipofuscinosis, neuronal, type 10	< 1 in 500	Reduced
11	CTSF	Ceroid lipofuscinosis, neuronal, type 13 (Kufs type)	N/A	N/A
1	CTSK	Pycnodysostosis	< 1 in 500	Reduced
10	CUBN	Megaloblastic anemia 1 (Imerslund-Grasbeck syndrome)	N/A	N/A
6	CUL7	3M syndrome 1	N/A	N/A
5	CWC27	Retinitis pigmentosa with or without skeletal anomalies	< 1 in 500	Reduced
10	CWF19L1	Spinocerebellar ataxia, autosomal recessive, type 17	N/A	N/A
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	N/A	N/A
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 7,143
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 407
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	N/A	N/A
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	N/A	N/A
4	CYP2U1	Spastic paraplegia, type 56, autosomal recessive	N/A	N/A

19	CYP4F22	Ichthyosis, congenital, autosomal recessive, type 5	N/A	N/A
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	N/A	N/A
1	DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	N/A	N/A
9	DBH	Dopamine beta-hydroxylase deficiency	N/A	N/A
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	N/A	N/A
11	DCHS1	Van Maldergem syndrome 1	N/A	N/A
10	DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabascan type	< 1 in 500	Reduced
11	DCPS	Al-Raqad syndrome	N/A	N/A
11	DDB2	Xeroderma pigmentosum, complementation group E	< 1 in 500	Reduced
7	DDC	Aromatic L-amino acid decarboxylase deficiency	N/A	N/A
14	DDHD1	Spastic paraplegia, type 28, autosomal recessive	N/A	N/A
8	DDHD2	Spastic paraplegia, type 54, autosomal recessive	< 1 in 500	Reduced
1	DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type	N/A	N/A
20	DDRGK1	Spondyloepimetaphyseal dysplasia, Shohat type	< 1 in 500	Reduced
12	DDX11	Warsaw breakage syndrome	N/A	N/A
1	DDX59	Orofaciodigital syndrome V	N/A	N/A
11	DENND5A	Epileptic encephalopathy, early infantile, 49	< 1 in 500	Reduced
2	DES	Myopathy, myofibrillar, type 1	N/A	N/A
8	DGAT1	?Diarrhea 7, protein-losing enteropathy type	N/A	N/A
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	N/A	N/A
19	DHPS	Neurodevelopmental disorder with seizures and speech and walking impairment	N/A	N/A
10	DHTKD1	2-aminoadipic 2-oxoadipic aciduria	N/A	N/A
5	DIAPH1	Seizures, cortical blindness, microcephaly syndrome	< 1 in 500	Reduced
2	DIS3L2	Perlman syndrome	N/A	N/A
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	N/A	N/A
5	DMGDH	Dimethylglycine dehydrogenase deficiency	N/A	N/A
4	DMP1	Hypophosphatemic rickets, autosomal recessive	< 1 in 500	Reduced
15	DMXL2	Developmental and epileptic encephalopathy, type 81	N/A	N/A
16	DNAAF1	Ciliary dyskinesia, primary, type 13	N/A	N/A
8	DNAAF11	Ciliary dyskinesia, primary, type 19	N/A	N/A
14	DNAAF2	Ciliary dyskinesia, primary, type 10	N/A	N/A
19	DNAAF3	Ciliary dyskinesia, primary, type 2	N/A	N/A
15	DNAAF4	Ciliary dyskinesia, primary, type 25	N/A	N/A
7	DNAAF5	Ciliary dyskinesia, primary, type 18	N/A	N/A
3	DNAH1	Spermatogenic failure, type 18	N/A	N/A
7	DNAH11	Ciliary dyskinesia, primary, type 7, with or without situs inversus	N/A	N/A
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	N/A	N/A
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	N/A
3	DNAJC19	3-methylglutaconic aciduria, type 5	< 1 in 500	Reduced
5	DNAJC21	Bone marrow failure syndrome, type 3	N/A	N/A
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	N/A	N/A
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	N/A	N/A
1	DOCK7	Epileptic encephalopathy, early infantile, 23	< 1 in 500	Reduced
9	DOCK8	Hyper-IgE recurrent infection syndrome, autosomal recessive	N/A	N/A
4	DOK7	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	1 in 262	1 in 719
9	DOLK	Congenital disorder of glycosylation, type 1M	< 1 in 500	<1 in 563
21	DONSON	Microcephaly, short stature, and limb abnormalities	N/A	N/A
11	DPAGT1	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	< 1 in 500	<1 in 808
17	DPH1	Developmental delay with short stature, dysmorphic features, and sparse hair	N/A	N/A
20	DPM1	Congenital disorder of glycosylation, type 1E	< 1 in 500	<1 in 1,750
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	N/A	N/A
1	DPYD	Dihydropyrimidine dehydrogenase deficiency	1 in 558	1 in 55,701
8	DPYS	Dihydropyrimidinuria	N/A	N/A
1	DRAM2	Cone-rod dystrophy 21	< 1 in 500	Reduced
2	DRC1	Ciliary dyskinesia, primary, type 21	N/A	N/A
18	DSG1	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE	< 1 in 500	Reduced
18	DSG4	Hypotrichosis, type 6	N/A	N/A
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	N/A	N/A
1	DSTYK	Spastic paraplegia, type 23, autosomal recessive	< 1 in 500	Reduced
6	DTNBP1	Hermansky-Pudlak syndrome, type 7	N/A	N/A
15	DUOX2	Thyroid dyshormonogenesis, type 6	1 in 80	1 in 170
15	DUOXA2	Thyroid dyshormonogenesis, type 5	1 in 80	1 in 144
18	DYM	Smith-McCort dysplasia; Dyggve-Melchior-Clausen disease	N/A	N/A
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	N/A	N/A
9	DYNC2I2	Short-rib thoracic dysplasia 11 with or without polydactyly	N/A	N/A
2	DYNC2LI1	Short-rib thoracic dysplasia 15 with polydactyly	N/A	N/A
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	N/A	N/A
16	EARS2	Combined oxidative phosphorylation deficiency 12	N/A	N/A
2	ECEL1	Arthrogryposis, distal, type 5D	N/A	N/A
10	ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	N/A	N/A
1	ECM1	Urbach-Wiethe disease	N/A	N/A
2	EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	1 in 131	1 in 252
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	N/A	N/A
7	EGFR	?Inflammatory skin and bowel disease, neonatal, 2	N/A	N/A
10	EGR2	Dejerine-Sottas disease	< 1 in 500	Reduced
2	EIF2AK3	Wolcott-Rallison syndrome	< 1 in 500	<1 in 2,500
15	EIF2AK4	Pulmonary venoocclusive disease 2	N/A	N/A
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	N/A	N/A
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	N/A	N/A
1	EMC1	Cerebellar atrophy, visual impairment, and psychomotor retardation	N/A	N/A
14	EML1	Band heterotopia	N/A	N/A
16	EMP2	Nephrotic syndrome, type 10	N/A	N/A
4	ENAM	Amelogenesis imperfecta, type 1C	N/A	N/A
17	ENO3	?Glycogen storage disease XIII	N/A	N/A
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	N/A	N/A
2	EPCAM	EPCAM-related conditions	< 1 in 500	Reduced
18	EPG5	Vici syndrome	N/A	N/A
6	EPM2A	Epilepsy, progressive myoclonic, type 2A (Lafora)	N/A	N/A
1	EPRS1	Leukodystrophy, hypomyelinating, type 15	N/A	N/A
11	EPS8L2	Deafness autosomal recessive, type 106	N/A	N/A
17	ERAL1	Perrault syndrome 6	< 1 in 500	Reduced
12	ERBB3	Lethal congenital contractural syndrome, type 2	N/A	N/A
19	ERCC1	Cerebrooculofacioskeletal syndrome, type 4	N/A	N/A
19	ERCC2	Trichothiodystrophy, type 1; Xeroderma pigmentosum, group D	1 in 500	1 in 10000
2	ERCC3	Trichothiodystrophy, type 2	1 in 436	1 in 1,306
16	ERCC4	Fanconi anemia, complementation group Q	N/A	N/A
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	N/A	N/A
5	ERCC8	Cockayne syndrome, type A	< 1 in 500	Reduced
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	N/A	N/A
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	N/A	N/A
8	EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities	N/A	N/A
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	N/A	N/A
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	N/A	N/A
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 1,000
15	FAN1	Interstitial nephritis, karyomegalic	N/A	N/A
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	N/A	N/A
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	N/A	N/A
2	FANCL	Fanconi anemia, complementation group L	N/A	N/A
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	N/A	N/A
2	FASTKD2	Combined oxidative phosphorylation deficiency 44	< 1 in 500	Reduced
4	FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2	N/A	N/A
14	FBLN5	Cutis laxa, autosomal recessive, type 1A	N/A	N/A
9	FBP1	Fructose-1,6-bisphosphatase deficiency	< 1 in 500	Reduced
6	FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	N/A	N/A
22	FBXO7	Parkinson disease, type 15, autosomal recessive	N/A	N/A
17	FDXR	Auditory neuropathy and optic atrophy	N/A	N/A
18	FECH	Protoporphyria, erythropoietic, autosomal recessive	N/A	N/A
20	FERMT1	Kindler syndrome	N/A	N/A
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	N/A	N/A
4	FGB	Congenital afibrinogenemia	< 1 in 500	Reduced
12	FGD4	Charcot-Marie-Tooth disease, type 4H	N/A	N/A
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	N/A	N/A
7	FKBP14	Ehlers-Danlos syndrome, kyphoscoliotic type, 2	N/A	N/A
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	N/A	N/A
11	FLI1	Bleeding disorder, platelet-type, type 21	< 1 in 500	Reduced
3	FLNB	Spondylocarpotarsal synostosis syndrome	N/A	N/A
1	FLVCR1	Posterior column ataxia-retinitis pigmentosa syndrome	N/A	N/A
14	FLVCR2	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	N/A	N/A
1	FMN2	Mental retardation, autosomal recessive, type 47	N/A	N/A
1	FMO3	Trimethylaminuria	1 in 100	1 in 1,000
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 822
4	FRAS1	Fraser syndrome, type 1	1 in 300	1 in 576
9	FREM1	Manitoba oculotrichoanal syndrome	N/A	N/A
13	FREM2	Fraser syndrome, type 2	N/A	N/A
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	N/A	N/A
21	FTCD	Glutamate formiminotransferase deficiency	1 in 355	1 in 591
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 1,149	1 in 4,880
14	FUT8	Congenital disorder of glycosylation with defective fucosylation, type 1	< 1 in 500	Reduced
9	FXN	Friedreich ataxia	1 in 91	1 in 1,014
3	FYCO1	Cataract 18	N/A	N/A
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 1,170
17	GAA	Glycogen storage disease, type 2	1 in 100	1 in 500
14	GALC	Krabbe disease	1 in 120	1 in 218
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 657
2	GALNT3	Tumoral calcinosis, hyperphosphatemic, familial, type 1	N/A	N/A
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	N/A	N/A
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 436
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	N/A	N/A
16	GCSH	Multiple mitochondrial dysfunctions syndrome 7	< 1 in 500	Reduced
8	GDAP1	Charcot-Marie-Tooth disease, recessive intermediate, type A	1 in 130	1 in 298
19	GDF1	Right atrial isomerism (Ivemark syndrome)	N/A	N/A
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	N/A	N/A
7	GHRHR	Growth hormone deficiency, isolated, type 1B	1 in 50	1 in 83
3	GHSR	Growth hormone deficiency, isolated partial	< 1 in 500	Reduced
20	GINS1	Immunodeficiency, type 55	N/A	N/A
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 42,000
1	GJC2	Spastic paraplegia, type 44, autosomal recessive	< 1 in 500	Reduced
3	GLB1	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	1 in 277	1 in 2770
9	GLDC	Glycine encephalopathy	1 in 180	1 in 720
15	GLDN	Lethal congenital contracture syndrome 11	N/A	N/A
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	N/A	N/A
9	GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism	N/A	N/A
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	N/A	N/A
5	GM2A	GM2-gangliosidosis, AB variant	< 1 in 500	Reduced
2	GMPPA	Alacrima, achalasia, and mental retardation syndrome	N/A	N/A
3	GMPPB	Muscular dystrophy-dystroglycanopathy 14	N/A	N/A
3	GNAT1	Night blindness, congenital stationary, type 1G	N/A	N/A
1	GNAT2	Achromatopsia, type 4	N/A	N/A
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 24,951
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 17,522
16	GNPTG	Mucolipidosis III gamma	< 1 in 500	Reduced
4	GNRHR	Hypogonadotropic hypogonadism, type 7, without anosmia	1 in 500	1 in 2,097
12	GNS	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	< 1 in 500	Reduced
1	GORAB	Geroderma osteodysplasticum	N/A	N/A
17	GOSR2	Epilepsy, progressive myoclonic, type 6	N/A	N/A
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	N/A	N/A
3	GP9	Bernard-Soulier syndrome, type C	1 in 451	1 in 4510
8	GPAA1	Glycosylphosphatidylinositol biosynthesis defect 15	N/A	N/A
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	N/A	N/A
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	N/A	N/A
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 224	1 in 2,236
13	GRK1	Oguchi disease-2	N/A	N/A
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	N/A	N/A
4	GRXCR1	Deafness, autosomal recessive, type 25	N/A	N/A

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	N/A	N/A
17	GUCY2D	Leber congenital amaurosis, type 1	1 in 248	1 in 305
4	GUF1	?Epileptic encephalopathy, early infantile, 40	N/A	N/A
7	GUSB	Mucopolysaccharidosis, type 7	1 in 552	1 in 1,6531
3	GYG1	Polyglucosan body myopathy, type 2	N/A	N/A
19	GYS1	Glycogen storage disease, type 0, muscle	< 1 in 500	Reduced
12	GYS2	Glycogen storage disease, type 0, liver	N/A	N/A
20	GZF1	Joint laxity, short stature, and myopia	< 1 in 500	Reduced
1	H6PD	Cortisone reductase deficiency 1	N/A	N/A
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	N/A	N/A
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 670
10	HELLS	Immunodeficiency-centromeric instability-facial anomalies syndrome 4	< 1 in 500	Reduced
11	HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A	N/A	N/A
15	HERC1	Macrocephaly, dysmorphic facies, and psychomotor retardation	< 1 in 500	Reduced
15	HERC2	Mental retardation, autosomal recessive, type 38	N/A	N/A
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
6	HFE	Hemochromatosis, type 1	1 in 10	1 in 200
1	HFM1	Premature ovarian failure 9	N/A	N/A
3	HGD	Alkaptonuria	1 in 250	1 in 2500
7	HGF	Deafness, autosomal recessive, type 39	< 1 in 500	Reduced
8	HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	1 in 345	1 in 4313
2	HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency	N/A	N/A
11	HIKESHI	Leukodystrophy, hypomyelinating, type 13	< 1 in 500	Reduced
5	HINT1	Neuromyotonia and axonal neuropathy, autosomal recessive	N/A	N/A
1	HJV	Hemochromatosis, type 2A	< 1 in 500	Reduced
10	HK1	Charcot-Marie-Tooth disease, type 4G	N/A	N/A
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	N/A	N/A
22	HMOX1	Heme oxygenase-1 deficiency	1 in 500	N/A
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 3380
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	N/A	N/A
1	HPCA	Dystonia 2, torsion, autosomal recessive	< 1 in 500	Reduced
12	HPD	Tyrosinemia, type 3	< 1 in 500	Reduced
4	HPGD	Hypertrophic osteoarthropathy, primary, type 1 (pachydermoperiostosis)	N/A	N/A
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 375
22	HPS4	Hermansky-Pudlak syndrome, type 4	N/A	N/A
11	HPS5	Hermansky-Pudlak syndrome, type 5	N/A	N/A
10	HPS6	Hermansky-Pudlak syndrome, type 6	N/A	N/A
10	HPSE2	Urofacial syndrome, type 1	N/A	N/A
8	HR	Alopecia universalis; Atrichia with papular lesions	N/A	N/A
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 2,750
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 1,625
10	HTRA1	CARASIL syndrome	N/A	N/A
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	N/A	N/A
11	HYLS1	Hydrolethalus syndrome	1 in 500	1 in 714
9	IARS1	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy	N/A	N/A
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 2186
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	N/A	N/A
16	IFT140	Retinitis pigmentosa, type 80; Short-rib thoracic dysplasia 9 with or without polydactyly	N/A	N/A
2	IFT172	Short-rib thoracic dysplasia 10 with or without polydactyly	N/A	N/A
14	IFT43	Short-rib thoracic dysplasia 18 with polydactyly	N/A	N/A
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	N/A	N/A
12	IFT81	Short-rib thoracic dysplasia 19 with or without polydactyly	N/A	N/A
12	IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency	< 1 in 500	Reduced
15	IGF1R	Insulin-like growth factor I, resistance to	< 1 in 500	Reduced
16	IGFALS	Acid-labile subunit deficiency	N/A	N/A
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 4,000
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	N/A	N/A
5	IL12B	Immunodeficiency, type 29, mycobacteriosis	< 1 in 500	Reduced
19	IL12RB1	Immunodeficiency, type 30	N/A	N/A
22	IL17RA	Immunodeficiency, type 51	< 1 in 500	Reduced
3	IL17RC	Candidiasis, familial, 9	N/A	N/A
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	N/A	N/A
3	ILDR1	Deafness, autosomal recessive, type 42	N/A	N/A
8	IMPA1	Mental retardation, autosomal recessive 59	< 1 in 500	Reduced
3	IMPG2	Retinitis pigmentosa, type 56	N/A	N/A
9	INPP5E	Joubert syndrome, type 1	< 1 in 500	Reduced
17	INPP5K	Muscular dystrophy, congenital, with cataracts and intellectual disability	N/A	N/A
11	INPPL1	Opsismodysplasia	N/A	N/A
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	N/A	N/A
9	INVS	Nephronophthisis, type 2, infantile	N/A	N/A
3	IQCB1	Senior-Loken syndrome, type 5	N/A	N/A
7	IQCE	Polydactyly, postaxial, type A7	N/A	N/A
12	IRAK4	Immunodeficiency, type 67 (IRAK4 deficiency)	N/A	N/A
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	N/A	N/A
17	ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	N/A	N/A
2	ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis	N/A	N/A
12	ITGA7	Muscular dystrophy, congenital, due to ITGA7 deficiency	N/A	N/A

10	ITGA8	Renal hypodysplasia/aplasia 1	N/A	N/A
21	ITGB2	Leukocyte adhesion deficiency	< 1 in 500	Reduced
17	ITGB3	Glanzmann thrombasthenia	N/A	N/A
17	ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia	< 1 in 500	Reduced
2	ITGB6	Amelogenesis imperfecta, type 1H	N/A	N/A
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	N/A	N/A
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 732
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	N/A	N/A
16	KATNB1	Lissencephaly 6, with microcephaly	N/A	N/A
16	KATNIP	Joubert syndrome 26	N/A	N/A
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	N/A	N/A
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	N/A	N/A
6	KHDC3L	Hydatidiform mole, recurrent, type 2	< 1 in 500	Reduced
14	KIAA0586	Joubert syndrome 23; Short-rib thoracic dysplasia 14 with polydactyly	N/A	N/A
17	KIAA0753	?Orofaciodigital syndrome, type 15	N/A	N/A
7	KIAA1549	Retinitis pigmentosa, type 86	N/A	N/A
1	KIF14	Microcephaly 20, primary, autosomal recessive; ?Meckel syndrome 12	N/A	N/A
2	KIF1A	Neuropathy, hereditary sensory, type 2C; Spastic paraplegia, type 30, autosomal recessive	N/A	N/A
17	KIF1C	Spastic ataxia 2, autosomal recessive	N/A	N/A
15	KIF7	Acrocallosal syndrome; Joubert syndrome, type 12	N/A	N/A
10	KIFBP	Goldberg-Shprintzen megacolon syndrome	< 1 in 500	Reduced
19	KISS1R	Hypogonadotropic hypogonadism, type 8, with or without anosmia	N/A	N/A
20	KIZ	Retinitis pigmentosa 69	N/A	N/A
5	KLHL3	Pseudohypoaldosteronism, type 2D	N/A	N/A
3	KLHL40	Nemaline myopathy 8, autosomal recessive	N/A	N/A
2	KLHL41	Nemaline myopathy 9	N/A	N/A
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	N/A	N/A
15	KNL1	Microcephaly 4, primary, autosomal recessive	< 1 in 500	Reduced
19	KPTN	Mental retardation, autosomal recessive 41	N/A	N/A
22	KREMEN1	Ectodermal dysplasia 13, hair/tooth type	< 1 in 500	Reduced
17	KRT10	Epidermolytic hyperkeratosis	N/A	N/A
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	N/A	N/A
3	KY	Myopathy, myofibrillar, type 7	< 1 in 500	Reduced
2	KYNU	Vertebral, cardiac, renal, and limb defects syndrome, type 2	N/A	N/A
14	L2HGDH	L-2-hydroxyglutaric aciduria	< 1 in 500	Reduced
18	LAMA1	Poretti-Boltshauser syndrome	N/A	N/A
6	LAMA2	LAMA2-related muscular dystrophy	1 in 125	1 in 625
18	LAMA3	Epidermolysis bullosa, junctional 2A, intermediate;Epidermolysis bullosa, junctional 2B, severe;Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous	< 1 in 500	Reduced
7	LAMB1	Lissencephaly, type 5	N/A	N/A
3	LAMB2	Pierson syndrome; Nephrotic syndrome, type 5, with or without ocular abnormalities	N/A	N/A
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	N/A	N/A
22	LARGE1	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	1 in 123	1 in 287
4	LARP7	Alazami syndrome	N/A	N/A
5	LARS1	?Infantile liver failure syndrome 1 (ILFS1)	N/A	N/A
3	LARS2	Perrault syndrome, type 4	N/A	N/A

16	LAT	Immunodeficiency, type 52	< 1 in 500	Reduced
1	LBR	Greenberg skeletal dysplasia	N/A	N/A
6	LCA5	Leber congenital amaurosis, type 5	< 1 in 500	Reduced
16	LCAT	Familial LCAT deficiency; Fish-eye disease	N/A	N/A
1	LCK	?Immunodeficiency, type 22	N/A	N/A
2	LCT	Lactase deficiency, congenital	N/A	N/A
11	LDHA	Glycogen storage disease type 11	< 1 in 500	Reduced
19	LDLR	Hypercholesterolemia, familial, type 1	N/A	N/A
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	N/A	N/A
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 1,060
6	LHFPL5	Deafness, autosomal recessive, type 67	< 1 in 500	Reduced
9	LHX3	Pituitary hormone deficiency, combined, type 3	1 in 1,398	1 in 13980
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	N/A	N/A
19	LIM2	Cataract 19, multiple types	< 1 in 500	Reduced
15	LINS1	Mental retardation, autosomal recessive, type 27	N/A	N/A
10	LIPA	Lysosomal acid lipase deficiency	1 in 112	1 in 2240
19	LIPE	Lipodystrophy, familial partial, type 6	N/A	N/A
3	LIPH	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	1 in 500	1 in 2,995
10	LIPN	Ichthyosis, congenital, autosomal recessive 8	< 1 in 500	Reduced
2	LIPT1	Lipoyltransferase 1 deficiency	N/A	N/A
11	LIPT2	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities	N/A	N/A
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	N/A	N/A
3	LMOD3	Nemaline myopathy 10	< 1 in 500	Reduced
19	LONP1	CODAS syndrome	N/A	N/A
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	N/A	N/A
18	LPIN2	Majeed syndrome	N/A	N/A
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	N/A	N/A
1	LRIG2	Urofacial syndrome 2	N/A	N/A
4	LRIT3	Night blindness, congenital stationary (complete), 1F, autosomal recessive	< 1 in 500	Reduced
10	LRMDA	Albinism, oculocutaneous, type 7	N/A	N/A
2	LRP2	Donnai-Barrow syndrome	< 1 in 500	Reduced
11	LRP4	Cenani-Lenz syndactyly syndrome	N/A	N/A
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	N/A	N/A
14	LTBP2	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma	N/A	N/A
11	LTBP3	Dental anomalies and short stature	N/A	N/A
19	LTBP4	Cutis laxa, autosomal recessive, type 1C	N/A	N/A
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	N/A	N/A
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	N/A	N/A
18	MALT1	Immunodeficiency, type 12	< 1 in 500	Reduced
9	MAN1B1	Mental retardation, autosomal recessive, type 15	N/A	N/A
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	N/A	N/A
15	MAPKBP1	Nephronophthisis 20	N/A	N/A
17	MAPT	Supranuclear palsy, progressive atypical (parkinsonism syndrome)	N/A	N/A

12	MARS1	Interstitial lung and liver disease	N/A	N/A
2	MARS2	Spastic ataxia, type 3, autosomal recessive	< 1 in 500	Reduced
5	MARVELD2	Deafness, autosomal recessive, type 49	N/A	N/A
3	MASP1	3MC syndrome 1	N/A	N/A
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	N/A	N/A
3	MCCC1	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	1 in 353	1 in 7060
5	MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	1 in 204	1 in 4080
2	MCEE	Methylmalonyl-CoA epimerase deficiency	1 in 500	1 in 50,000
2	MCFD2	Combined deficiency of factor V and factor VIII, type 2	N/A	N/A
5	MCIDAS	Ciliary dyskinesia, primary, type 42	< 1 in 500	Reduced
21	MCM3AP	Peripheral neuropathy, autosomal recessive, with or without impaired intellectual development	N/A	N/A
8	MCM4	Immunodeficiency, type 54	< 1 in 500	Reduced
6	MCM9	Ovarian dysgenesis 4	N/A	N/A
19	MCOLN1	Mucolipidosis type 4	1 in 1,166	1 in 4,850
8	MCPH1	Microcephaly type 1, primary, autosomal recessive	1 in 500	1 in 8,333
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	N/A	N/A
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	N/A	N/A
19	MEGF8	Carpenter syndrome, type 2	N/A	N/A
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	N/A	N/A
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	N/A	N/A
13	MIPEP	Combined oxidative phosphorylation deficiency 31	N/A	N/A
3	MITF	COMMAD syndrome	N/A	N/A
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 1,000
4	MMAA	Methylmalonic aciduria, vitamin B12-responsive	1 in 677	1 in 4513
12	MMAB	Methylmalonic aciduria, vitamin B12-responsive, type cblB	< 1 in 500	Reduced
1	MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	1 in 170	1 in 2429
2	MMADHC	Homocystinuria, cblD type, variant 1	< 1 in 500	Reduced
3	MME	Charcot-Marie-Tooth disease, axonal, type 2T	N/A	N/A
11	MMP13	Metaphyseal dysplasia, Spahr type	N/A	N/A
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	N/A	N/A
6	MMUT	Methylmalonic aciduria, mut(0) type	1 in 135	1 in 3375
18	MOCOS	Xanthinuria, type 2	N/A	N/A
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	<1 in 667
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	N/A	N/A
15	MPI	Congenital disorder of glycosylation, type 1B	1 in 473	1 in 11825
6	MPIG6B	Thrombocytopenia, anemia, and myelofibrosis	N/A	N/A
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	N/A	N/A

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	N/A	N/A
10	MRPS16	Combined oxidative phosphorylation deficiency 2	< 1 in 500	Reduced
3	MRPS22	Combined oxidative phosphorylation deficiency type 5	N/A	N/A
16	MRPS34	Combined oxidative phosphorylation deficiency 32	N/A	N/A
5	MSH3	Familial adenomatous polyposis, type 4	N/A	N/A
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	N/A	N/A
15	MTFMT	Combined oxidative phosphorylation deficiency 15	N/A	N/A
14	MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	N/A	N/A
1	MTHFR	Homocystinuria due to MTHFR deficiency	1 in 1,119	1 in 11190
11	MTMR2	Charcot-Marie-Tooth disease, type 4B1	1 in 500	1 in 1,000
6	MTO1	Combined oxidative phosphorylation deficiency 10	N/A	N/A
1	MTR	Homocystinuria-megaloblastic anemia, cblG complementation type	< 1 in 500	<1 in 864
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	N/A	N/A
1	MUTYH	Adenomas, multiple colorectal	N/A	N/A
12	MVK	Mevalonic aciduria	1 in 286	1 in 2,261
12	MYBPC1	Lethal congenital contracture syndrome, type 4	N/A	N/A
3	MYD88	Immunodeficiency, type 68	N/A	N/A
17	MYH2	Proximal myopathy and ophthalmoplegia	N/A	N/A
9	MYMK	Carey-Fineman-Ziter syndrome	< 1 in 500	Reduced
17	MYO15A	Deafness, autosomal recessive, type 3	N/A	N/A
22	MYO18B	Klippel-Feil syndrome, type 4, autosomal recessive, with myopathy and facial dysmorphism	N/A	N/A
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	N/A	N/A
18	MYO5B	Microvillus inclusion disease	N/A	N/A
6	MYO6	Deafness, autosomal recessive, type 37	< 1 in 500	Reduced
11	MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	1 in 129	1 in 2580
10	MYPN	Nemaline myopathy, type 11, autosomal recessive	< 1 in 500	Reduced
2	NADK2	2,4-dienoyl-CoA reductase deficiency	1 in 500	N/A
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	N/A	N/A
9	NANS	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type	< 1 in 500	Reduced
11	NARS2	Combined oxidative phosphorylation deficiency 24	N/A	N/A
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	N/A	N/A
3	NBEAL2	Gray platelet syndrome	N/A	N/A
8	NBN	Nijmegen breakage syndrome	1 in 525	1 in 17500
11	NCAPD3	Microcephaly 22, primary, autosomal recessive	N/A	N/A
7	NCF1	Chronic granulomatous disease, type 1	1 in 343	1 in 1,027
1	NCF2	Chronic granulomatous disease, type 2	< 1 in 500	Reduced
22	NCF4	Chronic granulomatous disease, type 3	N/A	N/A
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 19640
8	NDUFAF6	Mitochondrial complex I deficiency, nuclear type 17	N/A	N/A
2	NDUFB3	Mitochondrial complex I deficiency, nuclear type 25	N/A	N/A
8	NDUFB9	Mitochondrial complex I deficiency, nuclear type 24	N/A	N/A
2	NDUFS1	Mitochondrial complex I deficiency, nuclear type 5	N/A	N/A

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	N/A	N/A
17	NEK8	Renal-hepatic-pancreatic dysplasia, type 2	N/A	N/A
14	NEK9	Lethal congenital contracture syndrome 10	N/A	N/A
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	N/A	N/A
2	NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	N/A	N/A
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 24,964
14	NIN	Seckel syndrome, type 7	N/A	N/A
5	NIPAL4	Ichthyosis, congenital, autosomal recessive, type 6	N/A	N/A
8	NKX2-6	Conotruncal heart malformations	N/A	N/A
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	N/A	N/A
19	NLRP7	Hydatidiform mole, recurrent, type 1	1 in 500	1 in 722
7	NME8	Ciliary dyskinesia, primary, type 6	N/A	N/A
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 250	1 in 500
18	NPC1	Niemann-Pick disease, type C1	1 in 163	1 in 652
14	NPC2	Niemann-pick disease, type C2	< 1 in 500	Reduced
2	NPHP1	Joubert syndrome type 4	1 in 418	1 in 825
3	NPHP3	Meckel syndrome type 7	< 1 in 500	Reduced
1	NPHP4	Nephronophthisis type 4	< 1 in 500	Reduced
19	NPHS1	Nephrotic syndrome, type 1	1 in 112	1 in 1400
1	NPHS2	Nephrotic syndrome, type 2	1 in 226	1 in 2260
9	NPR2	Acromesomelic dysplasia, Maroteaux type	N/A	N/A
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	N/A	N/A
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	N/A	N/A
16	NTHL1	Familial adenomatous polyposis, type 3	N/A	N/A
1	NTRK1	Insensitivity to pain, congenital, with anhidrosis	1 in 1,122	1 in 11220
14	NUBPL	Mitochondrial complex I deficiency, nuclear type 21	N/A	N/A
12	NUP107	Nephrotic syndrome, type 11	N/A	N/A
19	NUP62	Striatonigral degeneration, infantile	< 1 in 500	Reduced
16	NUP93	Nephrotic syndrome, type 12	N/A	N/A
10	OAT	Gyrate atrophy of choroid and retina	< 1 in 500	Reduced
2	OBSL1	3M syndrome 2	N/A	N/A
15	OCA2	Oculocutaneous albinism type 2	1 in 101	1 in 204
5	OCLN	Pseudo-TORCH syndrome, type 1	< 1 in 500	Reduced
19	ODAD1	Ciliary dyskinesia, primary, type 20	N/A	N/A
10	ODAD2	Ciliary dyskinesia, primary, type 23	N/A	N/A
19	ODAD3	Ciliary dyskinesia, primary, type 30	< 1 in 500	Reduced
3	OPA1	Behr syndrome	N/A	N/A
19	OPA3	3-methylglutaconic aciduria, type 3	< 1 in 500	Reduced
10	OPTN	Amyotrophic lateral sclerosis, type 12	N/A	N/A

12	ORAI1	Immunodeficiency, type 9	< 1 in 500	Reduced
1	ORC1	Meier-Gorlin syndrome, type 1	N/A	N/A
2	ORC4	Meier-Gorlin syndrome, type 2	N/A	N/A
16	ORC6	Meier-Gorlin syndrome, type 3	N/A	N/A
14	OSGEP	Galloway-Mowat syndrome 3	N/A	N/A
6	OSTM1	Osteopetrosis, autosomal recessive type 5	< 1 in 500	Reduced
16	OTOA	Deafness, autosomal recessive, type 22	1 in 500	1 in 1667
2	OTOF	Deafness, autosomal recessive, type 9	1 in 228	1 in 22,701
11	OTOG	Deafness, autosomal recessive, type 18B	N/A	N/A
12	OTOGL	Deafness, autosomal recessive, type 84B	N/A	N/A
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	N/A	N/A
3	P2RY12	Bleeding disorder, platelet-type, type 8	< 1 in 500	Reduced
1	P3H1	Osteogenesis imperfecta, type 8	1 in 567	1 in 1,447
3	P3H2	Myopia, high, with cataract and vitreoretinal degeneration	N/A	N/A
1	PADI6	Preimplantation embryonic lethality 2	N/A	N/A
12	PAH	Phenylketonuria	1 in 60	1 in 857
16	PALB2	PALB2-related conditions	N/A	N/A
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	N/A	N/A
1	PARK7	Parkinson disease, type 7, autosomal recessive, early-onset	< 1 in 500	Reduced
16	PARN	Dyskeratosis congenita, autosomal recessive 6	N/A	N/A
15	PATL2	Oocyte maturation defect 4	N/A	N/A
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	N/A	N/A
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	N/A	N/A
10	PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	1 in 497	1 in 1657
14	PCK2	PEPCK deficiency, mitochondrial	N/A	N/A
21	PCNT	Microcephalic osteodysplastic primordial dwarfism, type 2	N/A	N/A
5	PCSK1	Obesity with impaired prohormone processing	N/A	N/A
3	PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy	< 1 in 500	Reduced
6	PDE10A	Dyskinesia, limb and orofacial, infantile-onset	N/A	N/A
5	PDE6A	Retinitis pigmentosa type 43	1 in 500	1 in 863
4	PDE6B	Retinitis pigmentosa type 40	1 in 200	1 in 4000
10	PDE6C	Cone dystrophy type 4	N/A	N/A
17	PDE6G	Retinitis pigmentosa type 57	< 1 in 500	Reduced
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	N/A	N/A
19	PEPD	Prolidase deficiency	N/A	N/A
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 1,287	1 in 16088
1	PEX11B	?Peroxisome biogenesis disorder 14B	N/A	N/A
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	N/A	N/A
11	PGAP2	Hyperphosphatasia with mental retardation syndrome 3	N/A	N/A
17	PGAP3	Hyperphosphatasia with mental retardation syndrome 4	N/A	N/A
1	PGM1	Congenital disorder of glycosylation, type 1t	N/A	N/A
6	PGM3	Immunodeficiency, type 23	N/A	N/A
1	PHGDH	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	1 in 631	1 in 4207
16	PHKB	Glycogen storage disease, type 9B	N/A	N/A
16	PHKG2	Glycogen storage disease type 9c	N/A	N/A
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	N/A	N/A
13	PIBF1	Joubert syndrome 33	N/A	N/A
16	PIEZO1	Lymphedema, hereditary, type 3	N/A	N/A
18	PIEZO2	Arthrogryposis, distal, with impaired proprioception and touch	N/A	N/A
1	PIGC	Glycosylphosphatidylinositol biosynthesis defect 16	< 1 in 500	Reduced
4	PIGG	Mental retardation, autosomal recessive 53	N/A	N/A
17	PIGL	Zunich neuroectodermal syndrome	N/A	N/A
1	PIGM	Glycosylphosphatidylinositol deficiency	< 1 in 500	Reduced
18	PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome, type 1	N/A	N/A
9	PIGO	Hyperphosphatasia with mental retardation syndrome 2	N/A	N/A
20	PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3	N/A	N/A
1	PIGV	Hyperphosphatasia with mental retardation syndrome 1	N/A	N/A
17	PIGW	Glycosylphosphatidylinositol biosynthesis defect 11	N/A	N/A
4	PIGY	Hyperphosphatasia with mental retardation syndrome 6	< 1 in 500	Reduced
1	PINK1	Parkinson disease, type 6, early onset	N/A	N/A
19	PIP5K1C	Lethal congenital contractural syndrome, type 3	< 1 in 500	Reduced
2	PJVK	Deafness, autosomal recessive, type 59	N/A	N/A
7	PKD1L1	Heterotaxy, visceral, 8, autosomal	N/A	N/A
6	PKHD1	Polycystic kidney disease type 4	1 in 66	1 in 264
1	PKLR	Pyruvate kinase deficiency	1 in 160	1 in 3200
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)	N/A	N/A
10	PLCE1	Nephrotic syndrome, type 3	< 1 in 500	Reduced
3	PLD1	Cardiac valvular defect, developmental	N/A	N/A
8	PLEC	Epidermolysis bullosa simplex with muscular dystrophy	N/A	N/A
1	PLEKHG5	Charcot-Marie-Tooth disease, recessive intermediate, type C	N/A	N/A
6	PLG	Plasminogen deficiency, type I	< 1 in 500	Reduced
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 299
3	PLOD2	Bruck syndrome 2	N/A	N/A
7	PLOD3	Lysyl hydroxylase 3 deficiency	N/A	N/A
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	N/A	N/A
7	PMPCB	Multiple mitochondrial dysfunctions syndrome 6	N/A	N/A
19	PNKP	Microcephaly, seizures, and developmental delay; Ataxia-oculomotor apraxia 4; ?Charcot-Marie-Tooth disease, type 2B2	N/A	N/A
14	PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency	< 1 in 500	Reduced
6	PNPLA1	Ichthyosis, congenital, autosomal recessive, type 10	N/A	N/A
11	PNPLA2	Neutral lipid storage disease with myopathy	N/A	N/A
19	PNPLA6	Boucher-Neuhauser syndrome; Oliver-McFarlane syndrome; Spastic paraplegia, type 39, autosomal recessive	N/A	N/A
17	PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	1 in 1,107	1 in 3,983
2	PNPT1	Combined oxidative phosphorylation deficiency 13	N/A	N/A
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	N/A	N/A
15	POLG	POLG-related disorders	1 in 194	1 in 340
6	POLH	Xeroderma pigmentosum, variant type	<1 in 500	Reduced
6	POLR1C	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	1 in 1,021	1 in 1,659
13	POLR1D	Treacher Collins syndrome, type 2	< 1 in 500	Reduced
10	POLR3A	Leukodystrophy, hypomyelinating, type 7	N/A	N/A

12	POLR3B	Leukodystrophy, hypomyelinating, type 8	N/A	N/A
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 31500
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 1,708
14	POMT2	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	< 1 in 500	Reduced
8	POP1	Anauxetic dysplasia, type 2	N/A	N/A
7	POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	1 in 268	1 in 1,069
3	POU1F1	Pituitary hormone deficiency, combined, type 1	1 in 32	1 in 126
4	PPA2	Sudden cardiac failure, infantile	N/A	N/A
15	PPIB	Osteogenesis imperfecta, type 9	N/A	N/A
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 4880
17	PRCD	Retinitis pigmentosa, type 36	N/A	N/A
9	PRDM12	Neuropathy, hereditary sensory and autonomic, type VIII	N/A	N/A
4	PRDM5	Brittle cornea syndrome, type 2	N/A	N/A
1	PRDX1	Methylmalonic aciduria and homocystinuria, cblC type, digenic	< 1 in 500	Reduced
2	PREPL	Myasthenic syndrome, congenital, type 22	N/A	N/A
10	PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2	1 in 308	1 in 538
1	PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	N/A	N/A
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	N/A	N/A
2	PRKRA	Dystonia, type 16	< 1 in 500	Reduced
16	PRMT7	Short stature, brachydactyly, intellectual developmental disability, and seizures	N/A	N/A
2	PROC	Thrombophilia due to protein C deficiency, autosomal recessive	N/A	N/A
22	PRODH	Hyperprolinemia, type 1	N/A	N/A
4	PROM1	Retinitis pigmentosa, type 41	1 in 323	1 in 6460
5	PROP1	Pituitary hormone deficiency, combined, type 2	1 in 84	1 in 4200
3	PROS1	Thrombophilia due to protein S deficiency, autosomal recessive	N/A	N/A
6	PRPH2	Leber congenital amaurosis 18; Retinitis punctata albescens	N/A	N/A
1	PRRX1	Agnathia-otocephaly complex	< 1 in 500	Reduced
4	PRSS12	Mental retardation, autosomal recessive, type 1	N/A	N/A
2	PRSS56	Microphthalmia, isolated, type 6	N/A	N/A
1	PRUNE1	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies	< 1 in 500	Reduced
19	PRX	Charcot-Marie-Tooth disease, type 4F	N/A	N/A
10	PSAP	Combined SAP deficiency	< 1 in 500	Reduced
9	PSAT1	Neu-Laxova syndrome, type 2	N/A	N/A
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	N/A	N/A
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	N/A	N/A
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	N/A	N/A
1	PYCR2	Leukodystrophy, hypomyelinating, type 10	< 1 in 500	Reduced
14	PYGL	Glycogen storage disease, type 6	N/A	N/A
11	PYGM	McArdle disease	1 in 206	1 in 2060
12	PYROXD1	Myopathy, myofibrillar, type 8	N/A	N/A
3	QARS1	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	N/A	N/A
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	N/A	N/A
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	N/A	N/A
1	RAB3GAP2	Martsolf syndrome 1; Warburg micro syndrome 2	N/A	N/A
5	RAD50	Nijmegen breakage syndrome-like disorder	N/A	N/A
17	RAD51C	RAD51C-related conditions	N/A	N/A
11	RAG1	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	1 in 344	1 in 614
11	RAG2	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	1 in 1,925	1 in 19250
11	RAPSN	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	1 in 165	1 in 1650
3	RARB	Microphthalmia, syndromic 12	< 1 in 500	Reduced
5	RARS1	Leukodystrophy, hypomyelinating, type 9	N/A	N/A
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 159	1 in 475
18	RBBP8	Jawad syndrome; Seckel syndrome, type 2	N/A	N/A
20	RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency	< 1 in 500	Reduced
1	RBM8A	Thrombocytopenia-absent radius syndrome	N/A	N/A
10	RBP3	?Retinitis pigmentosa 66	N/A	N/A
10	RBP4	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome	< 1 in 500	Reduced
13	RCBTB1	Retinal dystrophy with or without extraocular anomalies	N/A	N/A
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	N/A	N/A
11	RDX	Deafness, autosomal recessive, type 24	< 1 in 500	Reduced
8	RECQL4	Baller-Gerold syndrome; RAPADILINO syndrome; Rothmund-Thomson syndrome	N/A	N/A
19	REEP6	Retinitis pigmentosa 77	< 1 in 500	Reduced
7	RELN	Lissencephaly 2 (Norman-Roberts type)	N/A	N/A
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	N/A	N/A
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	N/A	N/A
15	RLBP1	Bothnia retinal dystrophy; Fundus albipunctatus	< 1 in 500	Reduced
6	RMND1	Combined oxidative phosphorylation deficiency 11	N/A	N/A
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 7,333
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	N/A	N/A
7	RNF216	Gordon Holmes syndrome	< 1 in 500	Reduced
11	ROBO3	Gaze palsy, familial horizontal, with progressive scoliosis, type 1	N/A	N/A
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	N/A	N/A
1	RPE65	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy	1 in 366	1 in 4575
14	RPGRIP1	Leber congenital amaurosis, type 6	N/A	N/A
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)	N/A	N/A
21	RSPH1	Ciliary dyskinesia, primary, type 24	N/A	N/A
6	RSPH3	Ciliary dyskinesia, primary, type 32	< 1 in 500	Reduced
6	RSPH4A	Ciliary dyskinesia, primary, type 11	N/A	N/A
6	RSPH9	Ciliary dyskinesia, primary, type 12	N/A	N/A
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	N/A	N/A
18	RTTN	Microcephaly, short stature, and polymicrogyria with seizures	N/A	N/A
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	Reduced
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	N/A	N/A
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	N/A	N/A
7	SBDS	Shwachman-Diamond syndrome	1 in 224	1 in 804
22	SBF1	Charcot-Marie-Tooth disease, type 4B3	N/A	N/A
11	SBF2	Charcot-Marie-Tooth disease, type 4B2	N/A	N/A
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	N/A	N/A
2	SCN9A	Indifference to pain and autosomal recessive hereditary sensory neuropathy type 2D	N/A	N/A
12	SCNN1A	Pseudohypoaldosteronism, type 1	N/A	N/A
16	SCNN1B	Pseudohypoaldosteronism, type 1	< 1 in 500	Reduced
16	SCNN1G	Pseudohypoaldosteronism, type 1	< 1 in 500	Reduced
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 8,333
11	SCYL1	Spinocerebellar ataxia, autosomal recessive, type 21	N/A	N/A
1	SDCCAG8	Bardet-Biedl syndrome, type 16	N/A	N/A
5	SDHA	Mitochondrial respiratory chain complex II deficiency; Leigh syndrome	N/A	N/A
19	SDHAF1	Mitochondrial complex II deficiency	< 1 in 500	Reduced
12	SDR9C7	Ichthyosis, congenital, autosomal recessive 13	N/A	N/A
14	SEC23A	Craniolenticulosutural dysplasia	N/A	N/A
20	SEC23B	Dyserythropoietic anemia, congenital, type 2	N/A	N/A
4	SEC24D	Cole-Carpenter syndrome 2	N/A	N/A
9	SECISBP2	Thyroid hormone metabolism, abnormal	N/A	N/A
1	SELENON	Muscular dystrophy, rigid spine, type 1	N/A	N/A
1	SEMA4A	Cone-rod dystrophy, type 10; Retinitis pigmentosa, type 35	N/A	N/A
4	SEPSECS	Pontocerebellar hypoplasia, type 2D	< 1 in 500	Reduced
6	SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome (MEGDEL)	N/A	N/A
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	N/A	N/A
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	N/A	N/A
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	N/A	N/A
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 1,132	1 in 5,468
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	N/A	N/A
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)	N/A	N/A
5	SKIC3	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	1 in 634	1 in 951
13	SLC10A2	Bile acid malabsorption, primary	N/A	N/A
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 100	1 in 1250
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 1,785
9	SLC1A1	Dicarboxylic aminoaciduria	< 1 in 500	Reduced
2	SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	< 1 in 500	Reduced
11	SLC22A12	Hypouricemia, renal	N/A	N/A
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	N/A	N/A
22	SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria	N/A	N/A
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	N/A	N/A
4	SLC25A4	Mitochondrial DNA depletion syndrome, type 12B (cardiomyopathic type) AR	< 1 in 500	Reduced
5	SLC25A46	Neuropathy, hereditary motor and sensory, type VIB	N/A	N/A
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	N/A	N/A
9	SLC27A4	Ichthyosis prematurity syndrome	N/A	N/A
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	N/A	N/A
3	SLC2A2	Fanconi-Bickel syndrome	< 1 in 500	Reduced
4	SLC2A9	Hypouricemia, renal, type 2	N/A	N/A
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	N/A	N/A
4	SLC34A2	Pulmonary alveolar microlithiasis	N/A	N/A
9	SLC34A3	Hypophosphatemic rickets with hypercalciuria	N/A	N/A
6	SLC35A1	Congenital disorder of glycosylation, type 2F	< 1 in 500	<1 in 1,500
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	N/A	N/A
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 84
1	SLC45A1	Intellectual developmental disorder with neuropsychiatric features	N/A	N/A
5	SLC45A2	Albinism, oculocutaneous, type 4	< 1 in 500	<1 in 1,600
17	SLC46A1	Folate malabsorption, hereditary	< 1 in 500	Reduced
17	SLC4A1	Distal renal tubular acidosis	N/A	N/A
20	SLC4A11	Corneal endothelial dystrophy, autosomal recessive	1 in 295	1 in 1475
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	N/A	N/A
16	SLC5A2	Renal glucosuria	N/A	N/A
19	SLC5A5	Thyroid dyshormonogenesis, type 1	1 in 72	1 in 96
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	N/A	N/A

1	SLC6A9	Glycine encephalopathy with normal serum glycine	< 1 in 500	Reduced
3	SLC7A14	Retinitis pigmentosa 68	N/A	N/A
14	SLC7A7	Lysinuric protein intolerance	< 1 in 500	Reduced
19	SLC7A9	Cystinuria	1 in 42	1 in 48
5	SLC9A3	Diarrhea 8, secretory sodium, congenital	< 1 in 500	Reduced
3	SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive, type 2	N/A	N/A
13	SLITRK6	Deafness and myopia	< 1 in 500	Reduced
8	SLURP1	Meleda disease	< 1 in 500	Reduced
16	SLX4	Fanconi anemia, complementation group P	N/A	N/A
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 36	1 in 360
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	N/A	N/A
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	N/A	N/A
12	SP7	Osteogenesis imperfecta, type XII	< 1 in 500	Reduced
8	SPAG1	Ciliary dyskinesia, primary, type 28	N/A	N/A
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	N/A	N/A
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 183
5	SPINK1	Tropical calcific pancreatitis	< 1 in 500	Reduced
5	SPINK5	Netherton syndrome	N/A	N/A
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	N/A	N/A
11	SPTBN2	Spinocerebellar ataxia, autosomal recessive, type 14	N/A	N/A
19	SPTBN4	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness	N/A	N/A
5	SQSTM1	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	N/A	N/A
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	N/A	N/A
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	N/A	N/A
2	STAMBP	Microcephaly-capillary malformation syndrome	< 1 in 500	Reduced
8	STAR	Lipoid adrenal hyperplasia	1 in 1,147	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	N/A	N/A
11	STIM1	Immunodeficiency, type 10	< 1 in 500	Reduced
20	STK4	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	N/A	N/A
15	STRA6	Microphthalmia, isolated, with coloboma, type 8	N/A	N/A
17	STRADA	Polyhydramnios, megalencephaly, and symptomatic epilepsy	N/A	N/A
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	N/A	N/A
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)	N/A	N/A
10	SUFU	Joubert syndrome, type 32	< 1 in 500	Reduced

7	SUGCT	Glutaric aciduria, type 3	N/A	N/A
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	N/A	N/A
19	SYNE4	Deafness, autosomal recessive, type 76	N/A	N/A
21	SYNJ1	Epileptic encephalopathy, early infantile, 53	< 1 in 500	Reduced
1	SYT14	?Spinocerebellar ataxia, autosomal recessive, type 11	N/A	N/A
1	SZT2	Epileptic encephalopathy, early infantile, 18	N/A	N/A
12	TAC3	Hypogonadotropic hypogonadism, type 10, with or without anosmia	N/A	N/A
17	TACO1	Mitochondrial complex IV deficiency, nuclear type 8	< 1 in 500	Reduced
4	TACR3	Hypogonadotropic hypogonadism, type 11, with or without anosmia	N/A	N/A
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	N/A	N/A
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	N/A	N/A
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	N/A	N/A
6	TBC1D7	Macrocephaly/megalencephaly syndrome, autosomal recessive	< 1 in 500	Reduced
17	TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	N/A	N/A
1	TBCE	Encephalopathy, progressive, with amyotrophy and optic atrophy; Hypoparathyroidism-retardation-dysmorphism syndrome; Kenny-Caffey syndrome, type 1	N/A	N/A
4	TBCK	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	N/A	N/A
1	TBX15	Cousin syndrome	< 1 in 500	Reduced
1	TBX19	Congenital isolated adrenocorticotropic hormone deficiency	< 1 in 500	Reduced
7	TBXAS1	Ghosal syndrome	N/A	N/A
17	TCAP	Limb-girdle muscular dystrophy, type 7 (LGMD R7)	< 1 in 500	Reduced
11	TCIRG1	Osteopetrosis, autosomal recessive, type 1	1 in 399	1 in 7980
22	TCN2	Transcobalamin II deficiency	N/A	N/A
12	TCTN1	Joubert syndrome, type 13	N/A	N/A
12	TCTN2	Joubert syndrome, type 24; ?Meckel syndrome, type 8	N/A	N/A
10	TCTN3	Joubert syndrome 18; Orofaciodigital syndrome IV	N/A	N/A
14	TDP1	?Spinocerebellar ataxia, autosomal recessive with axonal neuropathy	N/A	N/A
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 1,946	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	N/A	N/A
16	TELO2	You-Hoover-Fong syndrome	N/A	N/A
4	TENM3	Microphthalmia, isolated, with coloboma 9	N/A	N/A
5	TERT	Dyskeratosis congenita, autosomal recessive, type 4	< 1 in 500	Reduced
8	TEX15	Spermatogenic failure, type 25	N/A	N/A
3	TF	Atransferrinemia	1 in 500	1 in 7,143
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	N/A	N/A
14	TGM1	Ichthyosis, congenital, autosomal recessive, type 1	1 in 186	1 in 1860
15	TGM5	Peeling skin syndrome, type 2	N/A	N/A
11	TH	Segawa syndrome, recessive	< 1 in 500	Reduced
16	THOC6	Beaulieu-Boycott-Innes syndrome	N/A	N/A
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	N/A	N/A
9	TJP2	Cholestasis, progressive familial intrahepatic 4; Hypercholanemia, familial 1	N/A	N/A
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	N/A	N/A
9	TMC1	Deafness, autosomal recessive, type 7	N/A	N/A
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	N/A	N/A
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	N/A	N/A
2	TMEM237	Joubert syndrome, type 14	N/A	N/A
14	TMEM260	Structural heart defects and renal anomalies syndrome	N/A	N/A
8	TMEM67	Meckel syndrome 3; COACH syndrome 1; Joubert syndrome 6; Nephronophthisis 11	1 in 147	1 in 2,940
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	N/A	N/A
21	TMPRSS3	Deafness, autosomal recessive, type 8/10	1 in 135	1 in 2700
22	TMPRSS6	Iron-refractory iron deficiency anemia	N/A	N/A
12	TMTC3	Lissencephaly 8	N/A	N/A
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	N/A	N/A
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	N/A	N/A
17	TOP3A	Microcephaly, growth restriction, and increased sister chromatid exchange 2	N/A	N/A
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	N/A	N/A
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 84
11	TPP1	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	1 in 266	1 in 1773
9	TPRN	Deafness, autosomal recessive, type 79	N/A	N/A
2	TRAF3IP1	Senior-Loken syndrome, type 9	N/A	N/A
3	TRAIP	Seckel syndrome, type 9	< 1 in 500	Reduced
4	TRAPPC11	Limb-girdle muscular dystrophy, type 18 (LGMD R18)	N/A	N/A
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	N/A	N/A
6	TRDN	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	1 in 51	1 in 71
6	TREM2	Nasu-Hakola disease	< 1 in 500	Reduced
3	TREX1	Aicardi-Goutieres syndrome, type 1	1 in 98	1 in 186
8	TRHR	Hypothyroidism, congenital, nongoitrous, type 7	N/A	N/A
4	TRIM2	Charcot-Marie-Tooth disease, type 2R	< 1 in 500	Reduced
9	TRIM32	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	1 in 226	1 in 376
17	TRIM37	Mulibrey nanism	< 1 in 500	Reduced
22	TRIOBP	Deafness, autosomal recessive, type 28	1 in 445	1 in 8900
14	TRIP11	Achondrogenesis, type 1A	N/A	N/A
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	N/A	N/A
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	N/A	N/A
22	TRMU	Liver failure, transient infantile	< 1 in 500	Reduced
3	TRNT1	Retinitis pigmentosa and erythrocytic microcytosis	N/A	N/A
15	TRPM1	Night blindness, congenital stationary (complete), type 1C, autosomal recessive	N/A	N/A
9	TRPM6	Familial hypomagnesemia with secondary hypocalcemia	N/A	N/A
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	N/A	N/A
19	TSEN34	Pontocerebellar hypoplasia type 2C	N/A	N/A

17	TSEN54	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	1 in 223	1 in 3,997
12	TSFM	Combined oxidative phosphorylation deficiency, type 3	< 1 in 500	Reduced
1	TSHB	Hypothyroidism, congenital, nongoitrous, type 4	1 in 62	1 in 306
14	TSHR	Hypothyroidism, congenital, nongoitrous, type 1	1 in 62	1 in 189
17	TTC19	Mitochondrial complex III deficiency, nuclear type 2	N/A	N/A
2	TTC21B	Short-rib thoracic dysplasia, type 4, with or without polydactyly	N/A	N/A
2	TTC7A	Gastrointestinal defects and immunodeficiency syndrome	N/A	N/A
14	TTC8	Bardet-Biedl syndrome, type 8	N/A	N/A
8	TTI2	Mental retardation, autosomal recessive, type 39	N/A	N/A
14	TTLL5	Cone-rod dystrophy 19	N/A	N/A
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	N/A	N/A
15	TUBGCP4	Microcephaly and chorioretinopathy, autosomal recessive, type 3	N/A	N/A
22	TUBGCP6	Microcephaly and chorioretinopathy, autosomal recessive, type 1	N/A	N/A
16	TUFM	Combined oxidative phosphorylation deficiency 4	< 1 in 500	Reduced
6	TULP1	Retinitis pigmentosa 14; Leber congenital amaurosis 15	N/A	N/A
8	TUSC3	Mental retardation, autosomal recessive, type 7	N/A	N/A
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	N/A	N/A
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 1,400
3	UBA5	Epileptic encephalopathy, early infantile, 44	N/A	N/A
1	UBE2T	Fanconi anemia, complementation group T	< 1 in 500	Reduced
15	UBE3A	Angelman syndrome	1 in 62	N/A
12	UBE3B	Kaufman oculocerebrofacial syndrome	N/A	N/A
15	UBR1	Johanson-Blizzard syndrome	N/A	N/A
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 5,496
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	N/A	N/A
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	N/A	N/A
10	UROS	Porphyria, congenital erythropoietic	N/A	N/A
16	USB1	Poikiloderma with neutropenia	N/A	N/A
11	USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	1 in 257	1 in 3671
17	USH1G	Usher syndrome, type 1G	< 1 in 500	Reduced
1	USH2A	Usher syndrome, type 2A; Retinitis pigmentosa 39	1 in 70	1 in 467
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	N/A	N/A
6	VARS1	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy	N/A	N/A
6	VARS2	Combined oxidative phosphorylation deficiency 20	N/A	N/A
12	VDR	Rickets, vitamin D-resistant, type 2A	N/A	N/A
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	N/A	N/A
15	VPS33B	Arthrogryposis, renal dysfunction and cholestasis, type 1	N/A	N/A
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 1,506	N/A
14	VRK1	Pontocerebellar hypoplasia, type 1A	< 1 in 500	Reduced
14	VSX2	Microphthalmia with coloboma 3; Isolated microphthalmia 2	1 in 1,337	1 in 8913
12	VWF	von Willibrand disease, type 3	N/A	N/A

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	N/A	N/A
8	WASHC5	Ritscher-Schinzel syndrome, type 1	N/A	N/A
4	WDR19	Nephronophthisis, type 13; Senior-Loken syndrome, type 8	N/A	N/A
2	WDR35	Cranioectodermal dysplasia 2	N/A	N/A
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	N/A	N/A
15	WDR72	Amelogenesis imperfecta, type 2A3 (hypomaturation type)	N/A	N/A
15	WDR73	Galloway-Mowat syndrome 1	N/A	N/A
17	WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome, type 2	N/A	N/A
7	WEE2	Oocyte maturation defect 5	N/A	N/A
4	WFS1	Wolfram syndrome, type 1	1 in 370	1 in 3700
9	WHRN	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	1 in 93	1 in 127
2	WIPF1	?Wiskott-Aldrich syndrome 2	< 1 in 500	Reduced
12	WNK1	Neuropathy, hereditary sensory and autonomic, type 2	N/A	N/A
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	N/A	N/A
3	WNT7A	Fuhrmann syndrome	< 1 in 500	Reduced
17	WRAP53	Dyskeratosis congenita, autosomal recessive, type 3	N/A	N/A
8	WRN	Werner syndrome	1 in 474	1 in 681
16	WWOX	Epileptic encephalopathy, early infantile, type 28; Spinocerebellar ataxia, autosomal recessive, type 12	N/A	N/A
2	XDH	Xanthinuria, type 1	N/A	N/A
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	N/A	N/A
5	XRCC4	Short stature, microcephaly, and endocrine dysfunction	N/A	N/A
16	XYLT1	Desbuquois dysplasia, type 2	N/A	N/A
17	XYLT2	Spondyloocular syndrome	N/A	N/A
12	YARS2	Myopathy, lactic acidosis, and sideroblastic anemia, type 2	N/A	N/A
1	YY1AP1	Grange syndrome	N/A	N/A
2	ZAP70	Autoimmune disease, multisystem, infantile-onset, type 2; Immunodeficiency, type 48	N/A	N/A
11	ZBTB16	Skeletal defects, genital hypoplasia, and mental retardation	N/A	N/A
6	ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome, type 2	N/A	N/A
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	N/A	N/A
3	ZMYND10	Ciliary dyskinesia, primary, type 22	N/A	N/A
11	ZNF408	Retinitis pigmentosa, type 72	N/A	N/A
16	ZNF423	Joubert syndrome, type 19	N/A	N/A
16	ZNF469	Brittle cornea syndrome, type 1	N/A	N/A
17	ZNHIT3	PEHO syndrome	< 1 in 500	Reduced
11	ZP1	Oocyte maturation defect, type 1	N/A	N/A

N/A: no data prevalence unknown