

CGT Personalized v5.4.19

Patient Information		Sample Information		Clinic Information	
Unique pat id.:	0257696	Sample type:	Blood	Clinic:	WeFIV
Patient name:		Date of draw:	29/04/2025	Doctor:	CAROLINA BOUTEILLER
Patient DOB:		Date of receipt:	07/05/2025		
Ethnic group:	Caucasian	Report date/time:	05/03/2026		
Indication:	No family history				

TEST RESULTS

POSITIVE

The individual is carrier of:

Congenital adrenal hyperplasia due to 21-hydroxylase deficiency

Gene :	CYP21A2	Allele:	Hom
DNA Change:	NM_000500.9:c.1360C>T	Inheritance:	AR
Protein change:	p.Pro454Ser	OMIM phenotype:	201910
Variant classification:	Pathogenic / Likely Pathogenic		

INTERPRETATION OF TEST RESULTS

The variant c.1360C>T was detected in homozygous state in the CYP21A2 gene. This variant is considered mild and is associated with non-classical congenital adrenal hyperplasia. In men, it may cause early beard growth, but no gonadal abnormalities have been reported and sperm counts are typically normal (New 2006). In women, it has been described in association with hirsutism, irregular menstruation, and polycystic ovaries. This variant will be transmitted to all offspring. To avoid the risk of having affected children, the partner or gamete donor must test negative for the genes in which variants were identified. Direct relatives may also be carriers of one of the variants detected in this test. This information is important because it may be clinically relevant for them and for their offspring.

For genes with a negative test result, the risk of having affected children for the corresponding disorders decreased significantly compared to the general population. This is still true when partner or gamete donor is carrier for one or more of the analyzed genes. However, due to test limitations occurring in any genetic test this low risk is not zero (see limitations section and informed consent).

TEST DESCRIPTION

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

COMMENTS

The language of the report has been modified.

TEST METHODOLOGY

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

TEST LIMITATIONS

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

Analytically, the CGT test does not cover all known monogenic diseases nor all disease-causing variants for each tested gene. The test does not include the analysis of conditions associated with mitochondrial DNA nor multifactorial nor digenic inheritance. The test does not detect large rearrangements (inversions, deletions and duplications more than 15 nucleotides), variants located in regulatory regions or intronic regions outside the +/-3bp cut off (except if otherwise indicated), or in low sequence coverage areas (<7x). DNA changes caused by trinucleotide repeat expansions are not detected, except those indicated in the methodology section. For copy number variation analysis, when a normal result is obtained (2 copies detected), it is not possible to confirm that one copy is present in each of the two alleles (non-carrier) or if both copies are present in cis on the same allele, with no copies in the other allele (silent carrier). Clinical sensitivity varies among conditions. In particular, the sensitivity for SMN1 is approximately 96% because it is not possible to identify silent carriers among patients with 2 SMN1 copies detected and because point mutations or small indels are not analyzed. The CYP21A2 gene analysis presents unique challenges due to its high sequence homology (~98%) with the pseudogene CYP21A1P, which leads to frequent gene rearrangements and complex mutations. These challenges can cause difficulties in distinguishing CYP21A2 from CYP21A1P, increasing the risk of misdiagnosis. Different testing methods have specific limitations, requiring a combination of techniques such as long-range PCR, Sanger sequencing, next-generation sequencing (NGS), MLPA, and qPCR to achieve accurate results. These challenges and limitations may lead to false or inconclusive results. Therefore, genetic counselling is strongly recommended to evaluate the findings, discuss potential implications, and determine whether additional testing (such as MLPA) is necessary for an accurate diagnosis. In summary, sensitivity to detect pathogenic variants, if they result from complex gene conversion/gene rearrangements events, may be reduced. For the HEXB gene, the common 16 kb deletion that causes disease in 30% of affected patients is not included in CGT analysis. Furthermore, this test does not evaluate the HFE gene.

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

LEGAL/QUALITY

IGENOMIX ARGENTINA S.A will only release the report once a completed test requisition form is received. The clinic/clinician/certified health professional requesting the test is responsible for obtaining and taking custody of "Informed Consent" from the patient as depicted by national guidelines and/or legislation. This test was developed, and its performance characteristics determined by IGENOMIX SPAIN LAB, SLU. It has not been cleared or approved by the US Food and Drug Administration. The test is used as a laboratory developed test for clinical purposes.

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

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

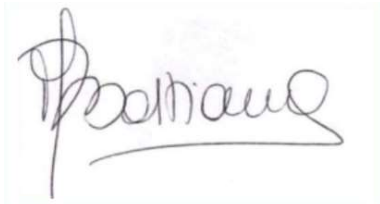
EXEMPTION CLAUSE OF DIAGNOSTIC LIABILITY

The genetic diagnosis services carried out by IGENOMIX ARGENTINA S.A are exclusively intended to be interpreted by qualified/certified health professionals.

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

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

SIGNED



Martina Di Bastiano

Laboratory Leader

COUNTERSIGNED



Arantxa Hervas PhD

3025-CV
Biotechnologist

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

Congenital adrenal hyperplasia due to 21-hydroxylase deficiency

What is Congenital adrenal hyperplasia due to 21-hydroxylase deficiency?

Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency (21-OHD) follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the CYP21A2 gene. This condition is associated with impaired hormone production by the adrenal glands, leading to excess androgen (or male sex hormones). The symptoms of 21-OHD CAH range in severity. There are two forms of 21-OHD: classic CAH and non-classic CAH. The combination of CYP21A2 variants may help to predict clinical presentation, with the inheritance of one or two mild variants in the CYP21A2 gene most often leading to non-classic CAH.

Classic 21-OHD CAH presents in infancy and is further divided into salt-wasting and simple virilizing types. With the salt-wasting form, affected males and females are unable to retain proper sodium (salt) levels, which can be life-threatening if left untreated. Infants with the simple virilizing form of 21-OHD CAH do not experience salt-wasting. In both types of classic 21-OHD CAH, female infants may be born with ambiguous genitalia. Males and females with classic 21-OHD CAH may show signs of precocious puberty, advanced bone age, and decreased fertility, and benefit from ongoing management.

Non-classic 21-OHD CAH is milder and more common; salt-wasting and ambiguous genitalia are not seen in individuals with the non-classic form of CAH. The onset of non-classic 21-OHD CAH symptoms is typically in adolescence, with variable degrees of postnatal androgen excess, leading to acne, excess hair growth, frontal balding, shorter stature in adulthood, and decreased fertility. Females with non-classic 21-OHD CAH may have menstrual irregularity and symptoms that could mimic polycystic ovary syndrome. Some individuals with non-classic 21-OHD CAH remain asymptomatic.

What is the next step if I am a carrier of Congenital adrenal hyperplasia due to 21-hydroxylase deficiency?

If you are a carrier of Congenital adrenal hyperplasia due to 21-hydroxylase 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?

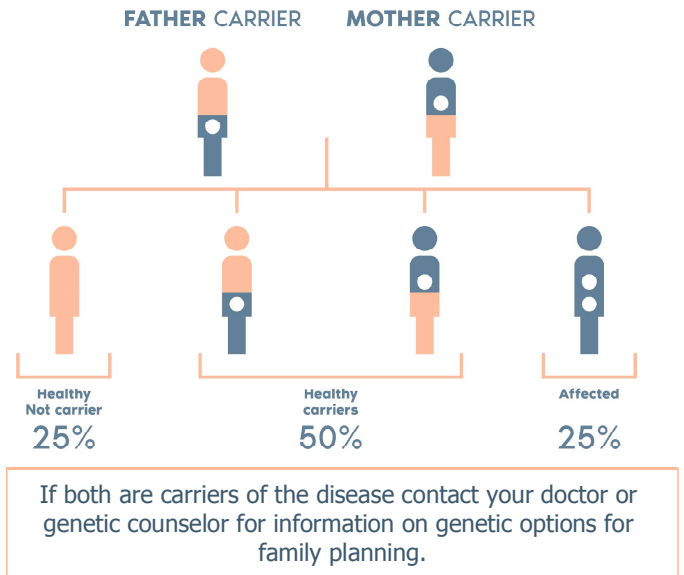
If your partner tests negative for Congenital adrenal hyperplasia due to 21-hydroxylase deficiency, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Congenital adrenal hyperplasia due to 21-hydroxylase deficiency?

When both parents are carriers of Congenital adrenal hyperplasia due to 21-hydroxylase 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.



LIST OF ANALYZED GENES

Total genes analyzed: 112. Genes analyzed AR: 97. Genes analyzed XL: 15.

Gene mean coverage >100x CHRNE, NAGA, CLCN1, CYP11A1, COL7A1, GJB2, ERCC2, FMO3, HBA1, HBA2, HBB, NEB, PRF1, ALPL, POLG, BBS1, DLD, BCKDHB, MVK, IDUA, DMD, OTC, GLA, F9, F8, L1CAM, FMR1, RPGR, LRP2, ABCC8, CPT2, ABCA3, PMM2, CFTR, SLC37A4, NPHS1, DHCR7, DYNC2H1, ELP1, SCO2, GRIP1, BLM, HPS1, CNGB3, MCOLN1, PCDH15, SLC26A4, MLC1, HPS3, BBS2, CLRN1, GBA1, CYP27A1, PKHD1, SLC26A2, GAA, ATP7B, TYR, GALT, ACADM, EVC2, FKTN, SMPD1, ACAT1, GBE1, GNPTAB, DHDDS, ASL, USH2A, AHI1, MCCC2, MMUT, CYP27B1, ACADVL, CEP290, RNASEH2B, CCDC88C, OCA2, RARS2, CC2D2A, PAH, ALDOB, XPC, AGA, TMEM216, CBS, ANO10, G6PC1, CYP21A2, FAH, FANCC

Gene mean coverage 50x-100x SLC6A8, ABCD1, ARX, PLP1, NR0B1, MID1, RS1, TNXB, AGXT, SLC19A3, HEXA, MCPH1, AIRE, ARSA, ASPA, MMACHC

Gene mean coverage < 50x TF, SMN1, FKRP, FXN, BTD

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	ARX	Epileptic encephalopathy, early infantile, type 1; ARX-related developmental disorders	1 in 25 000	1 in 100000
X	DMD	DMD-related conditions	1 in 2625	1 in 131250
X	F8	Hemophilia A	1 in 3500	1 in 89285
X	F9	Hemophilia B	1 in 6250	1 in 62500
X	FMR1	FMR1-related conditions	1 in 400	1 in 40000
X	GLA	Fabry disease	1 in 18750	1 in 187500
X	L1CAM	L1 Syndrome	1 in 7500	1 in 150000
X	MID1	Opitz GBBB syndrome, type 1	1 in 18750	1 in 125000
X	NROB1	Adrenal hypoplasia, congenital	1 in 17500	1 in 58333
X	OTC	Ornithine transcarbamylase deficiency	1 in 50000	1 in 166667
X	PLP1	Pelizaeus-Merzbacher disease	1 in 353	1 in 441
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	SLC6A8	Cerebral creatine deficiency syndrome, type 1	< 1 in 100 000	Reduced

Autosomal recessive conditions

Chrom	Gene	Disease/Condition	Carrier Rate	Residual Risk
16	ABCA3	Surfactant metabolism dysfunction, pulmonary, type 3	1 in 500	1 in 7,143
11	ABCC8	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	1 in 192	1 in 1920
1	ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	1 in 60	1 in 600
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
4	AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)	< 1 in 500	Reduced
2	AGXT	Hyperoxaluria, primary, type 1	1 in 174	1 in 2486
6	AHI1	Joubert syndrome, type 3	1 in 334	1 in 706
21	AIRE	Autoimmune polyendocrinopathy syndrome, type 1	1 in 310	1 in 4429
9	ALDOB	Fructose intolerance, hereditary	1 in 80	1 in 400
1	ALPL	ALPL-related conditions	1 in 274	1 in 2740
3	ANO10	Spinocerebellar ataxia, autosomal recessive, type 10	1 in 224	1 in 2,236
22	ARSA	Metachromatic leukodystrophy	1 in 192	1 in 1920
7	ASL	Argininosuccinic aciduria	1 in 116	1 in 1170
17	ASPA	Canavan disease	1 in 416	1 in 13867
13	ATP7B	Wilson disease	1 in 90	1 in 450
11	BBS1	Bardet-Biedl syndrome, type 1	1 in 152	1 in 1520
16	BBS2	Bardet-Biedl syndrome, type 2	1 in 200	1 in 4000
6	BCKDHB	Maple syrup urine disease, type 1B	1 in 365	1 in 2808
15	BLM	Bloom syndrome	1 in 320	1 in 3200
3	BTBD	Biotinidase deficiency	1 in 120	1 in 6000
21	CBS	Homocystinuria due to cystathionine beta-synthase	1 in 274	1 in 2740
4	CC2D2A	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2	1 in 196	1 in 2,800
14	CCDC88C	Hydrocephalus, congenital, type 1	1 in 500	1 in 7,143
12	CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	1 in 150	1 in 375
7	CFTR	Cystic fibrosis	1 in 25	1 in 833
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
7	CLCN1	Myotonia congenita, recessive	1 in 159	1 in 319
3	CLRN1	Usher syndrome, type 3A	1 in 250	1 in 1667
8	CNGB3	Achromatopsia, type 3	1 in 125	1 in 1250
3	COL7A1	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	1 in 150	1 in 1000
1	CPT2	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	1 in 100	1 in 667
15	CYP11A1	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	1 in 500	1 in 7,143
6	CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	1 in 62	1 in 1240
2	CYP27A1	Cerebrotendinous xanthomatosis	1 in 275	1 in 5500
12	CYP27B1	Vitamin D-dependent rickets, type 1	< 1 in 500	Reduced
11	DHCR7	Smith-Lemli-Opitz syndrome	1 in 100	1 in 1000
1	DHDDS	Retinitis pigmentosa, type 59	< 1 in 500	Reduced
7	DLD	Dihydroliipoamide dehydrogenase deficiency	< 1 in 500	Reduced
11	DYNC2H1	Short-rib thoracic dysplasia, type 3, with or without polydactyly	1 in 50	1 in 500
9	ELP1	Familial dysautonomia	1 in 200	1 in 2000
19	ERCC2	Trichothiodystrophy, type 1; Xeroderma pigmentosum, group D	1 in 500	1 in 10000
4	EVC2	Ellis-van Creveld syndrome	1 in 300	1 in 2000
15	FAH	Tyrosinemia, type 1	1 in 200	1 in 2000
9	FANCC	Fanconi anemia, complementation group C	1 in 480	1 in 2400
19	FKBP	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	FMO3	Trimethylaminuria	1 in 100	1 in 1,000
9	FXN	Friedreich ataxia	1 in 91	1 in 1,014
17	G6PC1	Glycogen storage disease, type 1A	1 in 300	1 in 3000
17	GAA	Glycogen storage disease, type 2	1 in 100	1 in 500
9	GALT	Galactosemia	1 in 109	1 in 727
1	GBA1	Gaucher Disease, type I-III; GD IIIC; GD, perinatal lethal	1 in 125	1 in 1563
3	GBE1	Glycogen storage disease, type 4	1 in 192	1 in 960
13	GJB2	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	1 in 40	1 in 500
12	GNPTAB	Mucopolipidosis 2 alpha/beta; Mucopolipidosis 3 alpha/beta	1 in 176	1 in 17,522
12	GRIP1	Fraser syndrome 3	1 in 224	1 in 2,236
16	HBA1	Alpha thalassemia	1 in 30	1 in 200
16	HBA2	Alpha thalassemia	1 in 30	1 in 200
11	HBB	HBB-related hemoglobinopathies	1 in 67	1 in 6700
15	HEXA	Tay-Sachs disease	1 in 250	1 in 1250
10	HPS1	Hermansky-Pudlak syndrome, type 1	1 in 493	1 in 4930
3	HPS3	Hermansky-Pudlak syndrome, type 3	1 in 300	1 in 1500
4	IDUA	Mucopolysaccharidosis type 1	1 in 153	1 in 3825
2	LRP2	Donnai-Barrow syndrome	< 1 in 500	Reduced

5	MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	1 in 204	1 in 4080
19	MCOLN1	Mucopolidosis 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
22	MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	< 1 in 500	Reduced
1	MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	1 in 170	1 in 2429
6	MMUT	Methylmalonic aciduria, mut(0) type	1 in 135	1 in 3375
12	MVK	Mevalonic aciduria	1 in 286	1 in 2,261
22	NAGA	Schindler disease, type I; Schindler disease, type III; Kanzaki disease	1 in 500	1 in 5000
2	NEB	Nemaline myopathy type 2	1 in 175	1 in 2188
19	NPHS1	Nephrotic syndrome, type 1	1 in 112	1 in 1400
15	OCA2	Oculocutaneous albinism type 2	1 in 101	1 in 1010
12	PAH	Phenylketonuria	1 in 60	1 in 857
10	PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	1 in 497	1 in 1657
6	PKHD1	Polycystic kidney disease type 4	1 in 66	1 in 264
16	PMM2	Congenital disorder of glycosylation, type 1A	1 in 71	1 in 3550
15	POLG	POLG-related disorders	1 in 194	1 in 3800
10	PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2	1 in 308	1 in 538
6	RARS2	Pontocerebellar hypoplasia, type 6	1 in 269	1 in 3363
13	RNASEH2B	Aicardi-Goutieres syndrome, type 2	1 in 440	1 in 7,333
22	SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1	1 in 500	1 in 8,333
2	SLC19A3	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)	1 in 232	1 in 1,785
5	SLC26A2	Achondrogenesis Ib; Atelosteogenesis, type II; De la Chapelle dysplasia; Diastrophic dysplasia; broad bone-platyspondylic variant; Epiphyseal dysplasia, multiple, 4	1 in 129	1 in 4300
7	SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome	1 in 88	1 in 587
11	SLC37A4	Glycogen storage disease, type 1B	1 in 500	1 in 7143
5	SMN1	Spinal muscular atrophy	1 in 50	1 in 588
11	SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B	1 in 350	1 in 3500
3	TF	Atransferrinemia	1 in 500	1 in 7,143
11	TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2	< 1 in 500	Reduced
6	TNXB	Ehlers-Danlos syndrome, classic-like	1 in 335	1 in 1675
11	TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	1 in 92	1 in 1840
1	USH2A	Usher syndrome, type 2A; Retinitis pigmentosa 39	1 in 60	1 in 600
3	XPC	Xeroderma pigmentosum, group C	< 1 in 500	Reduced

N/A: no data prevalence unknown