CGT Personalized v5.4.19

Patient Information		Sample Information		Clinic Information	
Unique pat id.:	0254840	**Sample type:**	Blood	**Clinic:**	WeFIV
Patient name:		**Date of draw:**	25/02/2025	**Doctor:**	CAROLINA BOUTEILLER
Patient DOB:		**Date of receipt:**	05/03/2025		
Ethnic group:	Unknown	**Report date/time:**	05/03/2026		
Indication:	No family history				

TEST RESULTS

POSITIVE

The individual is carrier of:

Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial

Gene :	COL7A1	**Allele:**	Het
DNA Change:	NM_000094.4:c.6502-2A>G	**Inheritance:**	AR
Protein change:	-	**OMIM phenotype:**	226600; 604129*; 131850*
Variant classification:	Pathogenic		

INTERPRETATION OF TEST RESULTS

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

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

For genes with a negative test result, the risk of having children affected by the associated disorders decreases significantly compared to the general population. This 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

Report's language has been updated by clinic's request.

TEST METHODOLOGY

DNA is isolated from the sample, usually blood or saliva, and analyzed by whole exome sequencing by NGS. This includes capture and sequence of all human exons and other gene regions of interest where known disease-causing variants are located. Sequencing raw data is then analyzed using bioinformatics (bioinformatic pipeline v3.0), which includes sequence alignment against the GRCh37 human genome reference, variant calling, annotation, and real-time interpretation of variants. QC parameters include, all reported samples that will have a minimum of 7Gb of data, with minimal mean coverage greater than 75x, and a specific depth analysis for more than 68,000 DNA positions where known pathogenic variants are located. In addition, complementary tests (non-NGS techniques) are performed for the following genes, if included, CFTR gene intronic variant/s; SMN1 gene exon 7-deletion; CYP21A2 gene frequent mutations; HBA1 and HBA2 genes frequent deletions; FXN gene GAA repeat sizing; FMR1gene CGG repeat sizing (females only); DMD gene frequent deletions/duplications; F8 gene intron 22 inversion (females only). When requested, CNV analysis by MLPA is performed for CFTR, HBB and HBA1/HBA2. Based on our validations studies, reported samples will have analytical detection rate for SNV variants as per the control sample NA12878 (Control positive); PASS value: NA12878 Sensitivity SNV ≥0.97000).

TEST LIMITATIONS

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

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

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

LEGAL/QUALITY

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

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

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

EXEMPTION CLAUSE OF DIAGNOSTIC LIABILITY

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

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

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

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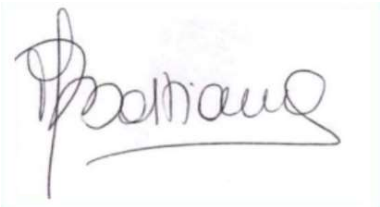
	
Martina Di Bastiano	**Arantxa Hervas PhD**
Laboratory Leader	3025-CV Biotechnologist

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

Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial

What is Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial?

Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type, follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the COL7A1 gene located on chromosomal region 3p21.1. The age of onset is neonatal/infantile. This disease is characterized by generalized cutaneous and mucosal blistering and scarring associated with severe deformities and major extracutaneous involvement. Some mutations may also cause a milder recessive form (non-HS type). Also, distint subclinical types of DEB include like Pruriginosa form (OMIM 604129), Pretibial form (OMIM 131850) and other are caused by mutations in this gene with variable modes of inheritace, including subclinical types caused by dominant mutations. In addition, less commonly, mutations in the COL7A1 gene may cause strictly dominant DEB. The overall prevalence of DEB is <1:1,000,000.

What is the next step if I am a carrier of Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial?

If you are a carrier of Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial 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?

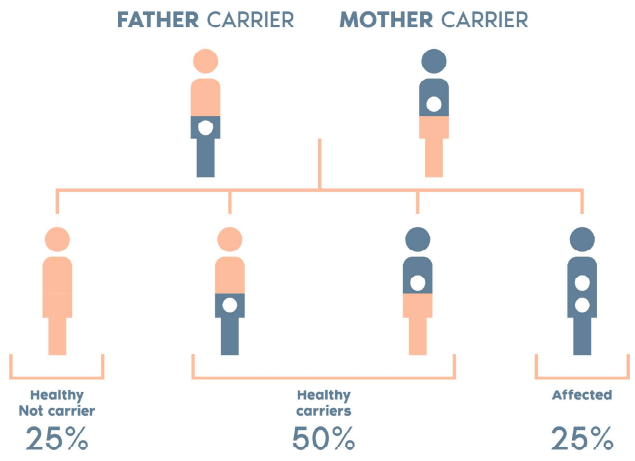
If your partner tests negative for Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there 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 Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial?

When both parents are carriers of Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

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



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

LIST OF ANALYZED GENES

Total genes analyzed: 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	NR0B1	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	BTD	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	Dihydrolipoamide 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	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	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	Mucolipidosis 2 alpha/beta; Mucolipidosis 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	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
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;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