

The information in this document relates to the new version of the CarrierTest, which is performed by the GNTlabs by GENNET laboratory starting December 1, 2025.

## Limitations, Methodology and Considerations

Like any laboratory test, CarrierTest has certain limitations. While it is a reliable screening method that provides valuable information on genetic carrier status, it cannot offer 100% certainty. The following considerations should be taken into account when interpreting the results.

### General Considerations and Possible Sources of Uncertainty

The test result is valid assuming the sample belongs to the tested individual. CarrierTest is performed under strict laboratory quality standards; however, as with any medical test, a very rare possibility of sample mix-up or technical issue during collection, labeling, or processing can never be completely excluded.

### Additional Information on Residual Risk

The CarrierTest is intended for healthy individuals who show no signs of a genetic disorder. The aim of the test is to reduce the risk of being a carrier, not to eliminate it entirely. The remaining (residual) risk of being a carrier after a negative CarrierTest result depends on the structure of the specific gene, the laboratory method used, and the carrier frequency in the European population, which may differ for individuals of other ethnic origins (see Table 2).

If a mutation is detected in one partner, the residual carrier risk of the other partner is used to calculate the risk of the offspring being affected.

The risk that both partners are carriers of a mutation in the same gene is very low after a negative CarrierTest result. For such a couple, the risk of having a child with an autosomal recessive disorder is therefore very low.

The risks of diseases arising from new mutations in germ cells or multigenic mutations may not be detected by this test.

### Technical and Analytical Aspects

CarrierTest uses whole-exome sequencing (WES), which analyzes a virtual panel of the (primarily) coding regions of selected human DNA genes (refer to Table 2) with overlaps into introns up to 50 bp. This test is based on massive parallel sequencing technology using short reads (SBS sequencing, Illumina), which is primarily suitable for analyzing SNP and small InDel variants. The sequencing data for each sample is subject to quality control, which ensures that all evaluable samples will have a minimum of 55M paired-end reads (clusters) after the removal of optical and PCR duplicates. The technical parameters of this method do not guarantee 100% coverage of all target regions.

Detecting certain variants or gene parts may not be possible due to local sequence characteristics, high/low genomic complexity, or the presence of closely related pseudogenes. Variants in promoter or deep intronic regions (unless specified otherwise), repetitive expansions (trinucleotide, hexanucleotide, or other), structural variants like inversions and gene conversions, and low-level mosaic variants may not be detected by this technology.

With the exception of specified genes or regions (see Table 1), CarrierTest does not analyze changes in the number of copies of genes or their parts (CNVs). The test is focused on germline mutations (mutations in germ cells). Somatic mutations are not examined. The test analyzes DNA and therefore does not investigate possible interactions between different genes or epigenetic factors.

For samples with lower quality (e.g., blood from patients with hematological disorders or highly degraded DNA), the quality of the NGS data may be reduced, which can lower the method's sensitivity for variant detection.

### Variant Classification and Pathogenicity

CarrierTest is a screening method that only detects selected pathogenic variants of classes 4/5 (pathogenic/likely pathogenic). Variant pathogenicity is evaluated based on current scientific and clinical knowledge (ClinVar/ClinSign databases) and may change over time. Should the classification of a detected variant change, GNTlabs will inform about this fact and offer an updated interpretation. The test cannot rule out mutations in other (uncovered, unanalyzed, or unevaluated) genes. GNTlabs, at its own discretion and in line with its commitment to the highest quality results, verifies NGS findings using complementary methods such as Sanger sequencing, long-range PCR, fragment analysis, or MLPA and StripAssay methods.

### Conclusion

CarrierTest is an internally developed and validated diagnostic tool accredited according to the standard ČSN EN ISO 15189:2013. Despite meeting the highest scientific and analytical standards, a residual risk cannot be excluded. Consultation of



the results with a qualified geneticist is beneficial, as they will consider the aforementioned limitations, as well as the family history, clinical picture, and all available information.

## Suggestion

The results of CarrierTest findings could be reviewed with a clinical geneticist, who may interpret the results for the patient and suggest possible treatment, monitoring, and preventive measures for the patient and their family.

**Table 1: Notes on the analysis of selected genes**

Gene	Notes
AR	The current testing method does not evaluate CAG trinucleotide repeat expansions in this gene.
CFTR	Analysis of the polymorphic region of intron 8 (e.g., the IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21.
CYP21A2	The CarrierTest is only a screening method that detects common variants of the <i>CYP21A2</i> gene: c.293-13C>G; p.Arg357Trp and p.Val282Leu, which is associated with a milder phenotype. Due to the presence of a highly homologous pseudogene and gene rearrangements in the corresponding genomic region, this method does not detect the presence of a chimeric gene (occurs in 30% of patients), changes in the copy number of parts of <i>CYP21A2</i> or the entire gene, the frequent p.Ile173Asn mutation (11% of patients), and other pathogenic mutations of the <i>CYP21A2</i> gene.
DMD	Single exon deletion/duplication analysis is limited to regions repeatedly published in the UMD database ( <a href="http://www.umd.be/DMD/W_DMD/">http://www.umd.be/DMD/W_DMD/</a> ).
GBA	The current testing method may not be able to reliably detect certain pathogenic variants in the <i>GBA</i> gene due to homologous recombination between the pseudogene and the functional gene.
HBB	The test is optimized for the detection of small variants (SNVs and indels) in coding regions and adjacent intron-exon boundaries. Large deletions/duplications may not be reliably detected by this method due to high sequence homology and the presence of segmental duplications. For these types of variants, complementary testing using MLPA is recommended.
HFE	Common variants c.187C>G (H63D) and c.193A>T (S65C) are associated with low penetrance and are not reported.
SERPINA1	The carrier test focuses on the Glu366Lys mutation (P1*Z mutation) (rs28929474). Homozygotes for the Glu366Lys mutation (ZZ genotype) account for 95% of patients with AAT deficiency with a reduction in AAT production to 15% of normal values. Other <i>SERPINA1</i> gene mutations are mostly significant only in combination with the "Z" mutation (compound heterozygote). In compound heterozygotes, AAT production is reduced to 30%-50% of normal values.
SMN1	The current testing method detects sequence variants in exon 7 and copy number variations in exons 7-8 of the <i>SMN1</i> gene (NM_022874.2). Sequencing and deletion/duplication analysis are not performed on any other regions in this gene. Approximately 5% to 8% of the population has two copies of <i>SMN1</i> on one chromosome and a deletion on the other, which is known as a [2+0] configuration (PubMed: 20301526). The current testing method cannot directly detect carriers with an <i>SMN1</i> [2+0] configuration, but it can detect the linkage between a silent carrier allele and certain population-specific single nucleotide changes. As a result, a negative carrier test result significantly reduces, but does not eliminate, the chance that a person is a carrier.
TYR	Due to interference from highly homologous regions, our current testing method has reduced sensitivity for detecting variants in exons 4-5 of the <i>TYR</i> gene (NM_000372.5).

**Table 2: List of genes analyzed within CarrierTest, associated diseases and residual risks**

Gene (RefSeq)	Disease/Syndrome	Ethnicity	Sensitivity	Carrier Rate	Residual Risk
ABCA3	ABCA3 Deficiency (ABCA3-related Pulmonary Surfactant Metabolism Dysfunction)	Caucasians	99%	1 in 100	1 in 10 000
		Finns	99%	1 in 500	1 in 50 000
		Ashkenazi	>95%	< 1 in 500	< 1 in 10 000
ABCC8	Congenital Hyperinsulinism (ABCC8-related)	Caucasians	99%	1 in 300	1 in 30 000
		Finns	99%	1 in 220	1 in 22 000



		Ashkenazi	99%	1 in 75	1 in 7500
<i>ABCD1</i>	Adrenoleukodystrophy, X-linked	General population (female)	99%	~1 in 20000	N/A
<i>ACADM</i>	Medium Chain Acyl-Coenzyme A Dehydrogenase (MCAD) Deficiency	Caucasians	99%	1 in 75	1 in 7 500
		Finns	99%	1 in 400	1 in 40 000
		Ashkenazi	99%	1 in 100	1 in 10 000
<i>ACADS</i>	Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency	Caucasians	99%	1 in 110	1 in 11 000
		Finns	99%	<1 in 500	<1 in 50 000
		Ashkenazi	99%	1 in 20	1 in 2000
<i>ACADVL</i>	Very Long Chain Acyl-Coenzyme A Dehydrogenase (VLCAD) Deficiency	Caucasians	99%	1 in 120	1 in 12 000
		Finns	99%	1 in 260	1 in 26 000
		Ashkenazi	99%	< 1 in 500	< 1 in 50 000
<i>ACAT1</i>	Beta-ketothiolase deficiency	General population	99%	<1 in 500	< 1 in 50 000
		Ashkenazi	97%	<1 in 500	<1 in 17 000
<i>ADGRV1</i>	Usher Syndrome Type 2C	Caucasians	98%	1 in 170	1 in 8 500
		Finns	98%	1 in 400	1 in 20 000
		Ashkenazi	98%	1 in 300	1 in 15 000
<i>AGA</i>	Aspartylglucosaminuria	General population	98%	< 1 in 500	1 in 25 000
		Finns	99%	1 in 36	1 in 3 500
<i>AGL</i>	Glycogen Storage Disease Type III	Caucasians	99%	1 in 160	1 in 16 000
		Finns	99%	<1 in 500	<1 in 50 000
		Ashkenazi	98%	<1 in 500	<1 in 25 000
<i>AGXT</i>	Primary Hyperoxaluria Type 1	Caucasians	97%	1 in 250	1 in 8 300
		Finns	99%	< 1 in 500	< 1 in 50 000
		Ashkenazi	99%	< 1 in 500	< 1 in 50 000
<i>AHI1</i>	Joubert Syndrome Type 3	Caucasians	99%	1 in 300	1 in 30 000
		Finns	99%	< 1 in 500	< 1 in 50 000
		Ashkenazi	99%	< 1 in 500	< 1 in 50 000
<i>AIRE</i>	Autoimmune Polyendocrinopathy with Candidiasis and Ectodermal Dysplasia	Caucasians	99%	1 in 150	1 in 15 000
		Finns	98%	1 in 90	1 in 4 500
		Ashkenazi	98%	<1 in 500	<1 in 25 000
<i>ALDOB</i>	Fructose Intolerance (Hereditary)	General population	99%	1 in 100	1 in 10 000
<i>ALPL</i>	Hypophosphatasia	Caucasians	99%	1 in 150	1 in 15 000
		Finns	99%	1 in 30	1 in 3 000
		Ashkenazi	99%	< 1 in 500	< 1 in 50 000
<i>ANO10</i>	Spinocerebellar Ataxia Type 10 (SCAR10)	General population	>95%	< 1 in 500	1 in 9981
<i>ANXA5</i>	N/A	General population	N/A	N/A	N/A
<i>AR</i>	Androgen Insensitivity Syndrome (X-linked)	General population (female)	70%	1 in 5000	N/A
<i>ARSA</i>	Metachromatic Leukodystrophy	General population	99%	1 in 100	1 in 10 000
<i>ASL</i>	Argininosuccinic Acid Lyase Deficiency	General population	99%	1 in 130	1 in 13 000



ASPA	Canavan Disease	General population	99%	1 in 150	1 in 15 000
		Ashkenazi	99%	1 in 55	1 in 5 000
ASS1	Citrullinemia	General population	99%	1 in 120	1 in 12 000
ATM	Ataxia-Telangiectasia	General population	>95%	1 in 166	1 in 3301
ATP7B	Wilson Disease	General population	99%	1 in 90	1 in 9 000
		Finns	99%	1 in 200	1 in 20 000
		Ashkenazi	99%	1 in 67	1 in 6 700
BBS1	Bardet-Biedl Syndrome Type 1	Caucasians	99%	1 in 150	1 in 15 000
		Finns	99%	1 in 300	1 in 30 000
		Ashkenazi	99%	< 1 in 500	< 1 in 50 000
BBS2	Bardet-Biedl Syndrome Type 2	General population	99%	< 1 in 500	< 1 in 50 000
		Ashkenazi	99%	1 in 130	1 in 13 000
BCKDHB	Maple Syrup Urine Disease Type 1B	General population	99%	< 1 in 500	< 1 in 50 000
		Finns	99%	1 in 175	1 in 17 500
		Ashkenazi	99%	1 in 75	1 in 7 500
BLM	Bloom Syndrome	Caucasians	99%	1 in 330	1 in 33 000
		Finns	99%	< 1 in 500	< 1 in 50 000
		Ashkenazi	98%	1 in 100	1 in 5 000
BTD	Biotinidase Deficiency	General population	99%	1 in 120	1 in 12 000
CBS	Homocystinuria	Caucasians	99%	1 in 150	1 in 15 000
		Finns	99%	1 in 500	1 in 50 000
		Ashkenazi		1 in 330	1 in 33 000
CC2D2A	Joubert Syndrome Type 9	General population	98%	1 in 200	1 in 10 000
CCDC88C	Congenital hydrocephalus 1	Caucasians	98%	<1 in 500	< 1 in 25,000
CDH23	Hearing loss, retinits	Caucasians	99%	1 in 250	1 in 4981
		Finns	99%	< 1 in 500	< 1 in 50 000
		Ashkenazi	99%	< 1 in 500	< 1 in 50 000
CEP290	Joubert syndrome 5, Leber congenital amaurosis 10	General population	99%	1 in 166	1 in 16 000
CFTR	Cystic fibrosis	General population	98%	1 in 24	1 in 1 150
CLCN1	Congenital myotonia, autosomal recessive form	Caucasians	99%	1 in 140	1 in 14 000
		Finns	99%	1 in 25	1 in 2 500
		Ashkenazi	99%	1 in 130	1 in 13 000
CLRN1	Usher Syndrome Type 3A	Caucasians	98%	1 in 460	1 in 23 000
		Finns	99%	1 in 70	1 in 7 000
		Ashkenazi	99%	1 in 95	1 in 9 500
CNGB3	Achromatopsia (CNGB3-related)	Caucasians	97%	1 in 120	1 in 4 000
		Finns	97%	1 in 170	1 in 5 600
		Ashkenazi	99%	1 in 270	1 in 27 000
COL4A5	Alport Syndrome (X-linked)	General population (female)	99%	1 in 5000	N/A



<i>COL7A1</i>	Epidermolysis Bullosa (Recessive Dystrophic)	General population	99%	1 in 160	1 in 16 000
<i>CPT2</i>	Carnitine Palmitoyltransferase II Deficiency	Caucasians	99%	1 in 180	1 in 18 000
		Finns	99%	1 in 240	1 in 24 000
		Ashkenazi	99%	1 in 40	1 in 4000
<i>CRB1</i>	Leber Congenital Amaurosis (CRB1-related)	General population	99%	1 in 320	1 in 32 000
<i>CTNS</i>	Cystinosis	General population	98%	< 1 in 500	<1 in 25 000
<i>CYP11A1</i>	Congenital Lipoid Adrenal Hyperplasia	N/A	99%	< 1 in 500	< 1 in 50,000
<i>CYP21A2</i>	Congenital Adrenal Hyperplasia (CYP21A2-related)	Caucasians	88%	1 in 70	1 in 580
		Ashkenazi	90%	1 in 40	1 in 390
<i>CYP27A1</i>	Cerebrotendinous Xanthomatosis (CTX)	General population	99%	1 in 300	1 in 30 000
<i>CYP27B1</i>	Vitamin D Dependent Rickets Type 1A	General population	99%	< 1 in 500	<1 in 50 000
<i>DHCR7</i>	Smith-Lemli-Opitz Syndrome	Caucasians	99%	1 in 50	1 in 5 000
		Finns	99%	1 in 181	1 in 18 000
		Ashkenazi	99%	1 in 40	1 in 4 000
<i>DHDDS</i>	Retinitis Pigmentosa 59	General population	99%	< 1 in 500	<1 in 50 000
		Ashkenazi	99%	1 in 120	1 in 12 000
<i>DLD</i>	Dihydrolipoamide Dehydrogenase Deficiency	General population	99%	< 1 in 500	<1 in 50 000
		Ashkenazi	99%	1 in 60	1 in 6 000
<i>DMD</i>	Duchenne and Becker Muscular Dystrophy (X-linked)	General population (female)	90%	1 in 3000	1 in 30000
<i>DNAJC30</i>	Leber Hereditary Optic Neuropathy (DNAJC30-related)	General population	N/A	N/A	N/A
<i>DYNC2H1</i>	Short-rib Polydactyly Syndrome Type III	General population	99%	< 1 in 500	<1 in 50 000
<i>ELP1</i>	Familial Dysautonomia	General population	99%	< 1 in 500	<1 in 50 000
		Ashkenazi	99%	1 in 37	1 in 3 700
<i>ERCC2</i>	Xeroderma pigmentosum	Caucasians	99%	1 in 200	1 in 20 000
		Finns	99%	< 1 in 500	<1 in 50 000
		Ashkenazi	99%	1 in 100	1 in 10 000
<i>EVC2</i>	Ellis-van Creveld Syndrome	General population	99%	< 1 in 500	<1 in 50 000
<i>F2</i>	Prothrombin Thrombophilia (F2-related)	General population	99%	1 in 150	1 in 14900
<i>F5</i>	Factor V Leiden Thrombophilia (F5-related)	General population	N/A	N/A	N/A
<i>F9</i>	Factor IX deficiency (hemophilia B)	General population (female)	99%	~1 in 20000	N/A
<i>FAH</i>	Tyrosinemia Type I	General population	99%	1 in 200	1 in 3981
<i>FANCA</i>	Fanconi Anemia Type A	General population	99%	1 in 200	1 in 20 000
<i>FANCC</i>	Fanconi Anemia Type C	General population	98%	< 1 in 500	1 in 25 000
		Ashkenazi	99%	1 in 89	1 in 9 000
<i>FKRP</i>	Limb-Girdle Muscular Dystrophy Type 2I	General population	99%	1 in 250	1 in 25 000
<i>FKTN</i>	Walker-Warburg Syndrome (FKTN-related)	General population	98%	< 1 in 500	1 in 25 000
		Ashkenazi	97%	1 in 80	1 in 2 600



<i>FMO3</i>	Trimethylaminuria	General population	99%	1 in 204	1 in 20 000
<i>G6PC</i>	Glycogen Storage Disease Type 1A	General population	99%	1 in 150	1 in 15 000
		Ashkenazi	99%	1 in 70	1 in 7 000
<i>GAA</i>	Pompe Disease	General population	99%	1 in 100	1 in 10 000
<i>GALT</i>	Galactosemia	General population	99%	1 in 84	1 in 8 400
<i>GBA</i>	Gaucher Disease	General population	>95%	1 in 137	<1 in 2 700
		Ashkenazi	>95%	1 in 15	<1 in 280
<i>GBE1</i>	Glycogen Storage Disease Type IV	General population	99%	1 in 300	1 in 30 000
		Ashkenazi	99%	1 in 70	1 in 7 000
<i>GCDH</i>	Glutaric Acidemia Type 1	General population	99%	1 in 150	1 in 15 000
<i>GJB2</i>	Non-Syndromic Hearing Loss/Deafness (GJB2-related)	General population	99%	1 in 30	1 in 3 000
		Ashkenazi	99%	1 in 13	1 in 1 2100
<i>GLA</i>	Fabry Disease (X-linked)	General population (female)	99%	< 1 in 500	N/A
<i>GLB1</i>	GM1 Gangliosidosis	General population	99%	1 in 220	1 in 22 000
<i>GLDC</i>	Glycine Encephalopathy (Nonketotic Hyperglycinemia)	General population	99%	1 in 333	1 in 33 000
<i>GNPTAB</i>	Mucolipidosis Type II/III	General population	98%	1 in 250	1 in 12 500
<i>GRIP1</i>	Fraser syndrome	Caucasians	99%	< 1 in 500	< 1 in 50,000
<i>HADHA</i>	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency	General population	99%	1 in 200	1 in 20 000
		Finns	99%	1 in 120	1 in 12000
<i>HBB</i>	Beta Thalassemia/Sickle Cell Disease	General population	99%	1 in 132	1 in 13 000
		Mediterranean	>95%	1 in 28	1 in 13 000
<i>HEXA</i>	Tay-Sachs Disease	General population	99%	1 in 200	1 in 20 000
		Ashkenazi	97%	1 in 30	1 in 581
<i>HFE</i>	Hemochromatosis (type I)	General population	100%	1 in 32	N/A
<i>HPS1</i>	Hermansky-Pudlak Syndrome Type 1	General population	98%	< 1 in 500	<1 in 25 000
<i>HPS3</i>	Hermansky-Pudlak Syndrome Type 3	General population	98%	< 1 in 500	1 in 25 000
		Ashkenazi	99%	1 in 250	1 in 25 000
<i>HSD17B4</i>	Peroxisomal Multifunctional Protein 2 Deficiency	General population	98%	< 1 in 500	1 in 25 000
<i>CHRNE</i>	Congenital Myasthenic Syndrome (CHRNE-related)	General population	>95%	< 1 in 500	<1 in 10 000
<i>CHST6</i>	Macular Corneal Dystrophy	General population	>95%	< 1 in 500	<1 in 10 000
<i>IDUA</i>	Mucopolysaccharidosis Type I (Hurler/Scheie Syndrome)	General population	99%	1 in 100	1 in 10 000
<i>IL2RG</i>	X-Linked Severe Combined Immunodeficiency (SCID)	General population (female)	99	1 in 25000	N/A
<i>KERA</i>	Cornea Planata 2	Finns	90%	1 in 122	1 in 1210
<i>L1CAM</i>	L1 Syndrome (X-linked)	General population (female)	>95%	N/A	N/A
<i>LRP2</i>	Donnai-Barrow Syndrome	Caucasians	98%	< 1 in 500	< 1 in 25,000
<i>MCCC1</i>	3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	General population	99%	1 in 120	1 in 12 000



<i>MCCC2</i>	3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	General population	99%	1 in 120	1 in 12 000
<i>MCOLN1</i>	Mucolipidosis Type IV	General population	97%	<1 in 500	<1 in 16 500
		Ashkenazi	97%	1 in 96	1 in 3 200
<i>MCPH1</i>	Microcephaly (Primary)	Caucasians	99%	1 in 416	1 in 42,000
<i>MEFV</i>	Familial Mediterranean Fever	General population	99%	1 in 250	1 in 25 000
		Mediterranean	99%	1 in 10	1 in 1 000
		Ashkenazi	99%	1 in 10	1 in 1 000
<i>MID1</i>	Opitz G/BBB Syndrome (X-linked)	General population (female)	90%	N/A	N/A
<i>MLC1</i>	Megalecephalic Leukoencephalopathy with Subcortical Cysts	General population	99%	< 1 in 500	<1 in 50 000
		Ashkenazi	99%	1 in 200	1 in 20 000
<i>MMACHC</i>	Methylmalonic Acidemia and Homocystinuria (CblC type)	General population	98%	1 in 170	1 in 8 500
<i>MMUT</i>	Methylmalonic Acidemia (Mut-related)	General population	99%	1 in 330	1 in 33 000
<i>MTM1</i>	Myotubular Myopathy (X-linked)	General population (female)	99%	1 in 25000	N/A
<i>MVK</i>	Mevalonate Kinase Deficiency	General population	99%	1 in 250	1 in 25 000
<i>MYO7A</i>	Usher Syndrome Type 1B	General population	99%	1 in 147	1 in 14 700
<i>NAGA</i>	Alpha-N-Acetylgalactosaminidase Deficiency (Schindler Disease)	Caucasians	99%	1 in 115	1 in 11 500
		Finns	99%	1 in 350	1 in 35 000
		Ashkenazi	99%	< 1 in 500	<1 in 50 000
<i>NBN</i>	Nijmegen Breakage Syndrome	General population	99%	< 1 in 500	<1 in 50 000
		Easter Europeans	99%	1 in 120	1 in 12 000
<i>NPC1</i>	Niemann-Pick Disease Type C1	General population	99%	1 in 200	1 in 20 000
<i>NPC2</i>	Niemann-Pick Disease Type C2	General population	99%	< 1 in 500	<1 in 50 000
<i>NPHS1</i>	Congenital Nephrotic Syndrome (NPHS1-related)	General population	99%	1 in 300	1 in 30 000
		Finns	97%	1 in 38	1 in 1234
		Ashkenazi	99%	< 1 in 500	<1 in 50 000
<i>OCA2</i>	Oculocutaneous Albinism Type 2	General population	>95%	< 1 in 500	1 in 9981
<i>OTC</i>	Ornithine Transcarbamylase (OTC) Deficiency (X-linked)	General population (female)	99%	1 in 20000	N/A
<i>PAH</i>	Phenylketonuria (PKU)	Caucasians	99%	1 in 40	1 in 981
		Finns	99%	1 in 170	1 in 17 000
		Ashkenazi	99%	1 in 17	1 in 1 700
<i>PCDH15</i>	Usher Syndrome Type 1F	General population	98%	1 in 400	1 in 20 000
		Ashkenazi	99%	1 in 116	1 in 11 600
<i>PEX1</i>	Zellweger Syndrome Spectrum (PEX1-related)	General population	98%	1 in 200	1 in 10 000
<i>PEX10</i>	Zellweger Syndrome Spectrum (PEX10-related)	General population	98%	< 1 in 500	<1 in 25 000
<i>PEX12</i>	Zellweger Syndrome Spectrum (PEX12-related)	General population	98%	< 1 in 500	<1 in 25 000



<i>PEX13</i>	Zellweger Syndrome Spectrum (PEX13-related)	General population	98%	< 1 in 500	<1 in 25 000
<i>PEX14</i>	Zellweger Syndrome Spectrum (PEX14-related)	General population	98%	< 1 in 500	<1 in 25 000
<i>PEX16</i>	Zellweger Syndrome Spectrum (PEX16-related)	General population	99%	< 1 in 500	<1 in 50 000
<i>PEX2</i>	Zellweger Syndrome Spectrum (PEX2-related)	General population	99%	< 1 in 500	<1 in 50 000
<i>PEX6</i>	Zellweger Syndrome Spectrum (PEX6-related)	General population	>95%	< 1 in 500	1 in 10 000
<i>PEX7</i>	Zellweger Syndrome Spectrum (PEX7-related)	General population	>95%	< 1 in 500	1 in 10 000
<i>PKHD1</i>	Autosomal Recessive Polycystic Kidney Disease (ARPKD)	General population	99%	1 in 70	1 in 7 000
		Finns	99%	1 in 38	1 in 3 700
<i>PLA2G6</i>	Neurodegeneration with Brain Iron Accumulation (PLA2G6-related)	General population	>95%	< 1 in 500	<1 in 10 000
<i>PMM2</i>	Congenital Disorder of Glycosylation Type Ia (CDG-Ia)	General population	99%	1 in 60	1 in 6 000
<i>POLG</i>	Progressive External Ophthalmoplegia (POLG-related)	General population	>95%	1 in 200	1 in 4 000
<i>PRF1</i>	Hemophagocytic Lymphohistiocytosis Type 2	General population	98%	< 1 in 500	<1 in 25 000
<i>RARS2</i>	Pontocerebellar hypoplasia	Caucasians	98%	1 in 364	1 in 18,000
<i>RNASEH2B</i>	Aicardi-Goutières Syndrome (RNASEH2B-related)	Caucasians	99%	1 in 195	1 in 19,000
<i>RS1</i>	Retinoschisis (X-linked)	General population (female)	>95%	< 1 in 500	1 in 9981
<i>SCO2</i>	Cytochrome c Oxidase Deficiency (SCO2-related)	Caucasians	98%	< 1 in 500	< 1 in 25,000
<i>SERPINA1</i>	Alpha-1 Antitrypsin Deficiency	General population	100%	1 in 32	N/A
<i>SGSH</i>	Mucopolysaccharidosis Type IIIA (Sanfilippo A)	General population	99%	1 in 230	1 in 23 000
<i>SLC19A3</i>	Biotin-Responsive Basal Ganglia Disease	General population	99%	< 1 in 500	<1 in 50 000
<i>SLC26A2</i>	Diastrophic Dysplasia	General population	99%	1 in 140	1 in 14 000
		Finns	99%	1 in 75	1 in 7 500
<i>SLC26A4</i>	Pendred Syndrome/Non-Syndromic Deafness (DFNB4)	General population	99%	1 in 75	1 in 7 500
<i>SLC37A4</i>	Glycogen Storage Disease Type Ib	General population	99%	< 1 in 500	<1 in 50 000
<i>SLC4A11</i>	Congenital Hereditary Endothelial Dystrophy (CHED)	General population	99%	< 1 in 500	<1 in 50 000
<i>SLC6A8</i>	Cerebral creatine deficiency syndromes	General population (female)	99%	~1 in 5000	N/A
<i>SMN1/SMN2</i>	Spinal Muscular Atrophy (SMA)	General population	100%	1 in 40	N/A
<i>SMPD1</i>	Niemann-Pick Disease Type A/B	General population	99%	< 1 in 500	<1 in 50 000
		Ashkenazi	99%	1 in 80	1 in 8 000
<i>TF</i>	Atransferrinemia	Caucasians	98%	< 1 in 500	< 1 in 25,000
<i>TGM1</i>	Congenital Ichthyosis (TGM1-related)	General population	99%	1 in 300	1 in 30 000



<i>TMEM216</i>	Joubert Syndrome Type 2	General population	99%	< 1 in 500	<1 in 50 000
		Ashkenazi	99%	1 in 110	1 in 11 000
<i>TPP1</i>	Neuronal Ceroid Lipofuscinosis Type 2 (CLN2)	General population	99%	1 in 180	1 in 18 000
<i>TYR</i>	Oculocutaneous Albinism Type 1	General population	99%	1 in 200	1 in 20 000
		Ashkenazi	99%	1 in 21	1 in 2 100
<i>UNC13D</i>	Hemophagocytic Lymphohistiocytosis Type 3	Caucasians	98%	1 in 293	1 in 15,000
<i>USH1C</i>	Usher Syndrome Type 1C	General population	98%	< 1 in 500	<1 in 25 000
		Ashkenazi	97%	1 in 235	1 in 7 800
<i>USH2A</i>	Usher Syndrome Type 2A	General population	99%	1 in 55	1 in 5 500
<i>XPC</i>	Xeroderma Pigmentosum Group C	General population	98%	< 1 in 500	<1 in 25 000

