RESULTS RECIPIENT
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Report Date: 01/28/2022

MALE
DONOR 10622
DOB:
Ethnicity: Northern European
Sample Type: EDTA Blood
Date of Collection:
Date Received: 01/22/2022
Date Tested: 01/28/2022
Barcode: 11004512969566
Accession ID: CSL9KFCXNLP3Z9P
Indication: Egg or sperm donor

FEMALE
N/A

Foresight® Carrier Screen

POSITIVE: CARRIER

ABOUT THIS TEST

The **Myriad Foresight Carrier Screen** utilizes sequencing, maximizing coverage across all DNA regions tested, to help you learn about your chance to have a child with a genetic disease.

RESULTS SUMMARY

Risk Details	DONOR 10622	Partner
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel **(175 conditions tested)**	N/A
POSITIVE: CARRIER Phenylalanine Hydroxylase Deficiency **Reproductive Risk: 1 in 200** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_000277.1(PAH):c.194T>C (I65T) heterozygote	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".

*Carriers generally do not experience symptoms.

No disease-causing mutations were detected in any other gene tested. A complete list of all conditions tested can be found on page 7.

CLINICAL NOTES

- None

NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner.
- Patients are recommended to discuss reproductive risks with their health care provider or a genetic counselor. Patients may also wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

POSITIVE: CARRIER

Phenylalanine Hydroxylase Deficiency

Reproductive risk: 1 in 200
Risk before testing: 1 in 9,500

Gene: PAH | **Inheritance Pattern:** Autosomal Recessive

Patient	DONOR 10622	No partner tested
Result	✚ Carrier	N/A
Variant(s)	NM_000277.1(PAH):c.194T>C(I65T) heterozygote	N/A
Methodology	Sequencing with copy number analysis (v3.1)	N/A
Interpretation	This individual is a carrier of phenylalanine hydroxylase deficiency. Carriers generally do not experience symptoms. The I65T mutation can be associated with any form of this disease.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000277:1-13.	N/A

What is Phenylalanine Hydroxylase Deficiency?

Phenylalanine hydroxylase deficiency (PAH deficiency), also called phenylketonuria (PKU), is an inherited disease in which the body cannot properly process the amino acid phenylalanine due to a deficient enzyme called phenylalanine hydroxylase. PAH deficiency is caused by mutations in the *PAH* gene. Phenylalanine is found in proteins and some other foods. If individuals with PAH deficiency do not get treatment, phenylalanine can accumulate to harmful levels, which can cause irreversible intellectual disability, seizures, developmental delay, and behavioral problems.

PAH deficiency causes a spectrum of disorders ranging from severe to nearly asymptomatic. The severity depends on the level of phenylalanine in the blood. It can be difficult to predict how severely affected a child will be based on the particular genetic mutations they carry. Children with any form of PAH deficiency should be evaluated by a specialist immediately after birth.

CLASSIC FORM

Classic PAH deficiency is the most common and severe form. Individuals with classic PAH deficiency produce little to no phenylalanine hydroxylase and are at risk for accumulating high levels of phenylalanine in their blood.

If PAH deficiency is not promptly diagnosed and treated with a special diet, intellectual disability will occur, along with a number of other symptoms including a small head, seizures, behavioral problems, a "mousy" or "musty" odor, abnormal gait, low bone density, and red, itchy skin (eczema). These are all avoidable if the proper diet is instituted shortly after birth and maintained throughout the lifespan.

MILD FORMS

Individuals who produce higher amounts of phenylalanine hydroxylase may have milder forms of PAH deficiency but are still at risk of developing the symptoms associated with classic PAH deficiency. Other names for the mild form include variant PKU or non-PKU hyperphenylalaninemia. Though the symptoms may be milder, there is still a risk for impaired mental development if the child's intake of phenylalanine is not monitored. Some individuals with mild PAH deficiency are able to tolerate a normal diet and do not require treatment. This will vary from person to person and must be determined by a medical professional based on the levels of phenylalanine in the person's blood.

How Common is Phenylalanine Hydroxylase Deficiency?

The prevalence of PAH deficiency is 1 in 10,000 in Caucasians, and it is more common in individuals of Turkish and Irish descent.

How is Phenylalanine Hydroxylase Deficiency Treated?

The degree of enzyme deficiency varies among people with PAH deficiency, and therefore the treatment must also be individualized based on the levels of phenylalanine in the blood. An infant with any form of PAH deficiency should be evaluated immediately after birth to determine whether or not he or she requires treatment. A blood test can reveal the amount of functioning phenylalanine hydroxylase in the body, and this will indicate the amount of phenylalanine the person can safely consume.

While individuals with classic PAH deficiency must adhere to a strict low-phenylalanine diet, others with milder forms can safely consume small amounts of the amino acid, and for some, treatment may not even be necessary.

Generally speaking, a diet low in protein and free from phenylalanine is important in preserving mental function in a person with classic PAH deficiency. Phenylalanine-free formulas are available for infants. Maintaining appropriate levels of phenylalanine in the brain can be achieved through blood testing and diet adjustment. This must be closely supervised by medical professionals. In most cases, this special diet must be maintained for life.

Individuals with any form of PAH deficiency should avoid consuming aspartame, an artificial sweetener containing phenylalanine.

Women with PAH deficiency who become pregnant must be particularly careful to maintain safe levels of phenylalanine in their bodies to avoid birth defects in their children. Ideally, this begins prior to conception.

In late 2007, the medication sapropterin dihydrochloride (brand name: Kuvan) was approved by the FDA to treat patients with PAH deficiency. For some patients, it can enhance the activity of the deficient enzyme and lower levels of phenylalanine in the body, allowing for a relaxation of the dietary restrictions. Some individuals with the disease do not respond to the drug; however, those who do respond to this treatment usually have milder forms of the disease.

What is the Prognosis for a Person with Phenylalanine Hydroxylase Deficiency?

If an individual with PAH deficiency is treated early and consistently, the prognosis can be excellent. Many with PAH deficiency have gone on to lead normal lives with normal intelligence and a normal lifespan. If treatment does not begin early or is not adequately maintained, a person with a more severe form of PAH deficiency is at risk for severe and irreversible brain damage.

Individuals with mild forms of PAH deficiency may lead a normal life without treatment.

Methods and Limitations

DONOR 10622 [Foresight Carrier Screen]: Sequencing with copy number analysis, spinal muscular atrophy, analysis of homologous regions, and alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (Assay(s): DTS v3.2).

Sequencing with copy number analysis

High-throughput sequencing and read-depth-based copy number analysis are used to analyze the genes listed in the Conditions Tested section of the report. Except where otherwise noted, the region of interest (ROI) comprises the indicated coding regions and 20 non-coding bases flanking each region. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected non-coding bases are excluded from the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. Select genes or regions for which pseudogenes or other regions of homology impede reliable variant detection may be assayed using alternate technology, or they may be excluded from the ROI. *CFTR* and *DMD* testing includes analysis for exon-level deletions and duplications with an average sensitivity of ~99%. Only exon-level deletions are assayed for other genes on the panel and such deletions are detected with a sensitivity of ≥75%. Selected founder deletions may be detected at slightly higher sensitivity. Affected exons and/or breakpoints of copy number variants are estimated from junction reads, where available, or using the positions of affected probes. Only exons known to be included in the region affected by a copy number variant are provided in the variant nomenclature. In some cases, the copy number variant may be larger or smaller than indicated. If *GJB2* is tested, large upstream deletions involving the *GJB6* and/or *CRYL1* genes that may affect the expression of *GJB2* are also analyzed.

Spinal muscular atrophy

Targeted copy number analysis via high-throughput sequencing is used to determine the copy number of exon 7 of the *SMN1* gene. Other genetic variants may interfere with this analysis. Some individuals with two copies of *SMN1* are "silent" carriers with both *SMN1* genes on one chromosome and no copies of the gene on the other chromosome. This is more likely in individuals who have two copies of the *SMN1* gene and are positive for the g.27134T>G single-nucleotide polymorphism (SNP) (PMID: 9199562, 23788250, and 28676062), which affects the reported residual risk; Ashkenazi Jewish or Asian patients with this genotype have a high post-test likelihood of being carriers for SMA and are reported as carriers. The g.27134T>G SNP is only reported in individuals who have two copies of *SMN1*.

Analysis of homologous regions

A combination of high-throughput sequencing, read-depth-based copy number analysis, and targeted genotyping is used to determine the number of functional gene copies and/or the presence of selected loss-of-function variants in certain genes that have homology to other genomic regions. The precise breakpoints of large deletions in these genes cannot be determined but are instead estimated from copy number analysis. Pseudogenes may interfere with this analysis, especially when many pseudogene copies are present.

If *CYP21A2* is tested, patients who have one or more additional copies of the *CYP21A2* gene and a pathogenic variant may or may not be a carrier of 21-hydroxylase deficient CAH, depending on the chromosomal location of the variants (phase). Benign *CYP21A2* gene duplications and/or triplications will only be reported in this context. Some individuals with two functional *CYP21A2* gene copies may be "silent" carriers, with two gene copies resulting from a duplication on one chromosome and a gene deletion on the other chromosome. This and other similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay. Given that the true incidence of non-classic CAH is unknown, the residual carrier and reproductive risk numbers on the report are based only on the published incidence for classic CAH. However, the published prevalence of non-classic CAH is highest in individuals of Ashkenazi Jewish, Hispanic, Italian, and Yugoslav descent. Therefore, the residual and reproductive risks are likely an underestimate for CAH, especially in the aforementioned populations, as they do not account for non-classic CAH.

Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis

High-throughput sequencing and read-depth-based copy number analysis are used to identify sequence variation and functional gene copies within the region of interest (ROI) of *HBA1* and *HBA2*, which includes the listed exons plus 20 intronic flanking bases. In a minority of cases where genomic features (e.g , long homopolymers) compromise calling fidelity, the affected intronic bases are not included in the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. For large deletions or duplications in these genes, the precise breakpoints cannot be determined but are instead estimated from copy number analysis. This assay has been validated to detect up to two additional copies of each alpha globin gene. In rare instances where assay results suggest greater than two additional copies are present, this will be noted but the specific number of gene copies observed will not be provided.

Extensive sequence homology exists between *HBA1* and *HBA2*. This sequence homology can prevent certain variants from being localized to one gene over the other. In these instances, variant nomenclature will be provided for both genes. If follow-up testing is indicated for patients with the nomenclature provided for both genes, both *HBA1* and *HBA2* should be tested. Some individuals with four functional alpha globin gene copies may be "silent" carriers, with three gene copies resulting from triplication on one chromosome and a single gene deletion on the other chromosome. This and other similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay.

Interpretation of reported variants

The classification and interpretation of all variants identified in this assay reflects the current state of Myriad's scientific understanding at the time this report was issued. Variants are classified according to internally defined criteria, which are compatible with the ACMG Standards and Guidelines for the Interpretation of Sequence Variants (PMID: 25741868). Variants that have been determined by Myriad to be disease-causing or likely disease-causing (i.e. pathogenic or likely pathogenic) are reported. Benign variants, variants of uncertain clinical significance (VUS), and variants not directly associated with the specified disease phenotype(s) are not reported. Variant classification and interpretation may change for a variety of reasons, including but not limited to, improvements to classification techniques, availability of additional scientific information, and observation of a variant in more patients. f the classification of one or more variants identified in this patient changes, an updated report reflecting the new classification generally will not be issued. If an updated report is issued, the variants reported may change based on their current classification. This can include changes to the variants displayed in gene specific 'variants tested' sections. Healthcare providers may contact Myriad directly to request updated variant classification information specific to this test result.

Limitations

The MWH Foresight Carrier Screen is designed to detect and report germline (constitutional) alterations. Mosaic (somatic) variation may not be detected, and if it is detected, it may not be reported. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes (phase). This test is not designed to detect sex-chromosome copy number variations. If present, sex-chromosome abnormalities may significantly reduce test sensitivity for X-linked conditions. Variant interpretation and residual and reproductive risk estimations assume a normal karyotype and may be different for individuals with abnormal karyotypes. The test does not fully address all inherited forms of intellectual disability, birth defects, or heritable diseases. Furthermore, not all forms of genetic variation are detected by this assay (i.e., duplications [except in specified genes], chromosomal rearrangements, structural abnormalities, etc.). Additional testing may be appropriate for some individuals. Pseudogenes and other regions of homology may interfere with this analysis. In an unknown number of cases, other genetic variation may interfere with variant detection. Rare carrier states where complementary changes exist between the chromosomes may not be detected by the assay. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions, and technical or analytical errors.

Detection rates are determined using published scientific literature and/or reputable databases, when available, to estimate the fraction of disease alleles, weighted by frequency, that the methodology is predicted to be able or unable to detect. Detection rates are approximate and only account for analytical sensitivity. Certain variants that have been previously described in the literature may not be reported, if there is insufficient evidence for pathogenicity. Detection rates do not account for the disease specific rates of *de novo* variation.

This test was developed, and its performance characteristics determined by, Myriad Women's Health, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's evaluation. CLIA Number: #05D1102604.

Incidental Findings

Unless otherwise indicated, these results and interpretations are limited to the specific disease panel(s) requested by the ordering healthcare provider. In some cases, standard data analyses may identify genetic findings beyond the region(s) of interest specified by the test, and such findings may not be reported. These findings may include genomic abnormalities with major, minor, or no, clinical significance.

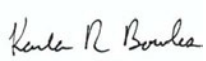
If you have questions or would like more information about any of the test methods or limitations, please contact (888) 268-6795.

Resources

GENOME CONNECT | http://www.genomeconnect.org

Patients can share their reports using research registries such as Genome Connect, an online research registry building a genetics and health knowledge base. Genome Connect provides patients, physicians, and researchers an opportunity to share genetic information to support the study of the impact of genetic variation on health conditions.

SENIOR LABORATORY DIRECTOR



Karla R. Bowles, PhD, FACMG, CGMB

Report content approved by Karla Bowles, PhD, FACMG, CGMB on Jan 28, 2022

Conditions Tested

6-pyruvoyl-tetrahydropterin Synthase Deficiency - **Gene:** PTS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000317:1-6. **Detection Rate:** Northern European >99%.

Adenosine Deaminase Deficiency - **Gene:** ADA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000022:1-12. **Detection Rate:** Northern European 98%.

Alpha Thalassemia, HBA1/HBA2-related - **Genes:** HBA1, HBA2. Autosomal Recessive. Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis. **Exons:** NM_000517:1-3; NM_000558:1-3. **Variants (16):** -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, Poly(A) AATAAA>AATA--, Poly(A) AATAAA>AATAAG, Poly(A) AATAAA>AATGAA, anti3.7, anti4.2, del HS-40. **Detection Rate:** Not calculated due to rarity of disease in this individual's reported ethnicity.

Alpha-mannosidosis - **Gene:** MAN2B1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000528:1-23. **Detection Rate:** Northern European >99%.

Alpha-sarcoglycanopathy - **Gene:** SGCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000023:1-9. **Detection Rate:** Northern European >99%.

Alstrom Syndrome - **Gene:** ALMS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015120:1-23. **Detection Rate:** Northern European >99%.

Andermann Syndrome - **Gene:** SLC12A6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_133647:1-25. **Detection Rate:** Northern European >99%.

Argininemia - **Gene:** ARG1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000045:1-8. **Detection Rate:** Northern European 97%.

Argininosuccinic Aciduria - **Gene:** ASL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001024943:1-16. **Detection Rate:** Northern European >99%.

Aspartylglucosaminuria - **Gene:** AGA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000027:1-9. **Detection Rate:** Northern European >99%.

Ataxia with Vitamin E Deficiency - **Gene:** TTPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000370:1-5. **Detection Rate:** Northern European >99%.

Ataxia-telangiectasia - **Gene:** ATM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000051:2-63. **Detection Rate:** Northern European 96%.

ATP7A-related Disorders - **Gene:** ATP7A. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000052:2-23. **Detection Rate:** Northern European 90%.

Autoimmune Polyglandular Syndrome Type 1 - **Gene:** AIRE. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000383:1-14. **Detection Rate:** Northern European >99%.

Autosomal Recessive Osteopetrosis Type 1 - **Gene:** TCIRG1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006019:2-20. **Detection Rate:** Northern European 96%.

Autosomal Recessive Polycystic Kidney Disease, PKHD1-related - **Gene:** PKHD1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_138694 2-67. **Detection Rate:** Northern European >99%.

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay - **Gene:** SACS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_014363 2-10. **Detection Rate:** Northern European 99%.

Bardet-Biedl Syndrome, BBS1-related - **Gene:** BBS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_024649:1-17. **Detection Rate:** Northern European >99%.

Bardet-Biedl Syndrome, BBS10-related - **Gene:** BBS10. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_024685:1-2. **Detection Rate:** Northern European >99%.

Bardet-Biedl Syndrome, BBS12-related - **Gene:** BBS12. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_152618:2. **Detection Rate:** Northern European >99%.

Bardet-Biedl Syndrome, BBS2-related - **Gene:** BBS2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_031885:1-17. **Detection Rate:** Northern European >99%.

BCS1L-related Disorders - **Gene:** BCS1L. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004328:3-9. **Detection Rate:** Northern European >99%.

Beta-sarcoglycanopathy - **Gene:** SGCB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000232:1-6. **Detection Rate:** Northern European >99%.

Biotinidase Deficiency - **Gene:** BTD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000060:1-4. **Detection Rate:** Northern European >99%.

Bloom Syndrome - **Gene:** BLM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000057:2-22. **Detection Rate:** Northern European >99%.

Calpainopathy - **Gene:** CAPN3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000070:1-24. **Detection Rate:** Northern European 99%.

Canavan Disease - **Gene:** ASPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000049:1-6. **Detection Rate:** Northern European 98%.

Carbamoylphosphate Synthetase I Deficiency - **Gene:** CPS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001875:1-38. **Detection Rate:** Northern European >99%.

Carnitine Palmitoyltransferase IA Deficiency - **Gene:** CPT1A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001876:2-19. **Detection Rate:** Northern European >99%.

Carnitine Palmitoyltransferase II Deficiency - **Gene:** CPT2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000098:1-5. **Detection Rate:** Northern European >99%.

Cartilage-hair Hypoplasia - **Gene:** RMRP. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NR_003051:1. **Detection Rate:** Northern European >99%.

Cerebrotendinous Xanthomatosis - **Gene:** CYP27A1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000784:1-9. **Detection Rate:** Northern European >99%.

Citrullinemia Type 1 - **Gene:** ASS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000050:3-16. **Detection Rate:** Northern European >99%.

CLN3-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001042432 2-16. **Detection Rate:** Northern European >99%.

CLN5-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006493:1-4. **Detection Rate:** Northern European >99%.

CLN8-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_018941:2-3. **Detection Rate:** Northern European >99%.

Cohen Syndrome - **Gene:** VPS13B. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017890:2-62. **Detection Rate:** Northern European 97%.

COL4A3-related Alport Syndrome - **Gene:** COL4A3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000091:1-52. **Detection Rate:** Northern European 94%.

COL4A4-related Alport Syndrome - **Gene:** COL4A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000092:2-48. **Detection Rate:** Northern European >99%.
Combined Pituitary Hormone Deficiency, PROP1-related - **Gene:** PROP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006261:1-3. **Detection Rate:** Northern European >99%.
Congenital Adrenal Hyperplasia, CYP11B1-related - **Gene:** CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000497:1-9. **Detection Rate:** Northern European 97%.
Congenital Adrenal Hyperplasia, CYP21A2-related - **Gene:** CYP21A2. Autosomal Recessive. Analysis of homologous regions. **Variants (13):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Northern European 96%.
Congenital Disorder of Glycosylation Type Ia - **Gene:** PMM2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000303:1-8. **Detection Rate:** Northern European >99%.
Congenital Disorder of Glycosylation Type Ic - **Gene:** ALG6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_013339:2-15. **Detection Rate:** Northern European >99%.
Congenital Disorder of Glycosylation, MPI-related - **Gene:** MPI. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002435:1-8. **Detection Rate:** Northern European >99%.
Costeff Optic Atrophy Syndrome - **Gene:** OPA3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_025136:1-2. **Detection Rate:** Northern European >99%.
Cystic Fibrosis - **Gene:** CFTR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection Rate:** Northern European >99%.
Cystinosis - **Gene:** CTNS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004937:3-12. **Detection Rate:** Northern European >99%.
D-bifunctional Protein Deficiency - **Gene:** HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000414:1-24. **Detection Rate:** Northern European 98%.
Delta-sarcoglycanopathy - **Gene:** SGCD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000337:2-9. **Detection Rate:** Northern European 96%.
Dihydrolipoamide Dehydrogenase Deficiency - **Gene:** DLD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000108:1-14. **Detection Rate:** Northern European >99%.
Dysferlinopathy - **Gene:** DYSF. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003494:1-55. **Detection Rate:** Northern European 98%.
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) - **Gene:** DMD. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_004006:1-79. **Detection Rate:** Northern European 99%.
ERCC6-related Disorders - **Gene:** ERCC6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000124:2-21. **Detection Rate:** Northern European 96%.
ERCC8-related Disorders - **Gene:** ERCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000082:1-12. **Detection Rate:** Northern European 97%.
EVC-related Ellis-van Creveld Syndrome - **Gene:** EVC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153717:1-21. **Detection Rate:** Northern European 96%.
EVC2-related Ellis-van Creveld Syndrome - **Gene:** EVC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_147127:1-22. **Detection Rate:** Northern European 98%.
Fabry Disease - **Gene:** GLA. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000169:1-7. **Detection Rate:** Northern European 98%.
Familial Dysautonomia - **Gene:** ELP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003640:2-37. **Detection Rate:** Northern European >99%.
Familial Hyperinsulinism, ABCC8-related - **Gene:** ABCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000352:1-39. **Detection Rate:** Northern European >99%.
Familial Hyperinsulinism, KCNJ11-related - **Gene:** KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000525:1. **Detection Rate:** Northern European >99%.
Familial Mediterranean Fever - **Gene:** MEFV. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000243:1-10. **Detection Rate:** Northern European >99%.
Fanconi Anemia Complementation Group A - **Gene:** FANCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000135:1-43. **Detection Rate:** Northern European 92%.
Fanconi Anemia, FANCC-related - **Gene:** FANCC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000136:2-15. **Detection Rate:** Northern European >99%.
FKRP-related Disorders - **Gene:** FKRP. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_024301:4. **Detection Rate:** Northern European >99%.
FKTN-related Disorders - **Gene:** FKTN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001079802:3-11. **Detection Rate:** Northern European >99%.
Free Sialic Acid Storage Disorders - **Gene:** SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012434:1-11. **Detection Rate:** Northern European 98%.
Galactokinase Deficiency - **Gene:** GALK1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000154:1-8. **Detection Rate:** Northern European >99%.
Galactosemia - **Gene:** GALT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000155:1-11. **Detection Rate:** Northern European >99%.
Gamma-sarcoglycanopathy - **Gene:** SGCG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000231:2-8. **Detection Rate:** Northern European 87%.
Gaucher Disease - **Gene:** GBA. Autosomal Recessive. Analysis of homologous regions. **Variants (10):** D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs*18. **Detection Rate:** Northern European 60%.
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness - **Gene:** GJB2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004004:1-2. **Detection Rate:** Northern European >99%.
GLB1-related Disorders - **Gene:** GLB1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000404:1-16. **Detection Rate:** Northern European >99%.
Glutaric Acidemia, GCDH-related - **Gene:** GCDH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000159:2-12. **Detection Rate:** Northern European >99%.
Glycine Encephalopathy, AMT-related - **Gene:** AMT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000481:1-9. **Detection Rate:** Northern European >99%.
Glycine Encephalopathy, GLDC-related - **Gene:** GLDC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000170:1-25. **Detection Rate:** Northern European 94%.
Glycogen Storage Disease Type Ia - **Gene:** G6PC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000151:1-5. **Detection Rate:** Northern European 98%.
Glycogen Storage Disease Type Ib - **Gene:** SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001164277 3-11. **Detection Rate:** Northern European >99%.
Glycogen Storage Disease Type III - **Gene:** AGL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000642:2-34. **Detection Rate:** Northern European >99%.
GNE Myopathy - **Gene:** GNE. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001128227:1-12. **Detection Rate:** Northern European >99%.
GNPTAB-related Disorders - **Gene:** GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_024312:1-21. **Detection Rate:** Northern European >99%.

HADHA-related Disorders - **Gene:** HADHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000182:1-20. **Detection Rate:** Northern European >99%.

Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - **Gene:** HBB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000518:1-3. **Detection Rate:** Northern European >99%.

Hereditary Fructose Intolerance - **Gene:** ALDOB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000035:2-9. **Detection Rate:** Northern European >99%.

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - **Gene:** HEXA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000520:1-14. **Detection Rate:** Northern European >99%.

HMG-CoA Lyase Deficiency - **Gene:** HMGCL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000191:1-9. **Detection Rate:** Northern European >99%.

Holocarboxylase Synthetase Deficiency - **Gene:** HLCS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000411:4-12. **Detection Rate:** Northern European >99%.

Homocystinuria, CBS-related - **Gene:** CBS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000071:3-17. **Detection Rate:** Northern European >99%.

Hydrolethalus Syndrome - **Gene:** HYLS1. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_145014:4. **Detection Rate:** Northern European >99%.

Hypophosphatasia - **Gene:** ALPL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000478:2-12. **Detection Rate:** Northern European >99%.

Isovaleric Acidemia - **Gene:** IVD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002225:1-12. **Detection Rate:** Northern European >99%.

Joubert Syndrome 2 - **Gene:** TMEM216. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001173990:1-5. **Detection Rate:** Northern European >99%.

Junctional Epidermolysis Bullosa, LAMA3-related - **Gene:** LAMA3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000227:1-38. **Detection Rate:** Northern European >99%.

Junctional Epidermolysis Bullosa, LAMB3-related - **Gene:** LAMB3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000228:2-23. **Detection Rate:** Northern European >99%.

Junctional Epidermolysis Bullosa, LAMC2-related - **Gene:** LAMC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005562:1-23. **Detection Rate:** Northern European >99%.

Krabbe Disease - **Gene:** GALC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000153:1-17. **Detection Rate:** Northern European >99%.

Leigh Syndrome, French-Canadian Type - **Gene:** LRPPRC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_133259:1-38. **Detection Rate:** Northern European >99%.

Lipoid Congenital Adrenal Hyperplasia - **Gene:** STAR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000349:1-7. **Detection Rate:** Northern European >99%.

Lysosomal Acid Lipase Deficiency - **Gene:** LIPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000235:2-10. **Detection Rate:** Northern European 98%.

Maple Syrup Urine Disease Type Ia - **Gene:** BCKDHA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000709:1-9. **Detection Rate:** Northern European >99%.

Maple Syrup Urine Disease Type Ib - **Gene:** BCKDHB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_183050:1-10. **Detection Rate:** Northern European >99%.

Maple Syrup Urine Disease Type II - **Gene:** DBT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001918:1-11. **Detection Rate:** Northern European 97%.

Medium Chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADM. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000016:1-12. **Detection Rate:** Northern European >99%.

Megalencephalic Leukoencephalopathy with Subcortical Cysts - **Gene:** MLC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015166 2-12. **Detection Rate:** Northern European >99%.

Metachromatic Leukodystrophy - **Gene:** ARSA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000487:1-8. **Detection Rate:** Northern European >99%.

Methylmalonic Acidemia, cblA Type - **Gene:** MMAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_172250:2-7. **Detection Rate:** Northern European >99%.

Methylmalonic Acidemia, cblB Type - **Gene:** MMAB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_052845:1-9. **Detection Rate:** Northern European >99%.

Methylmalonic Acidemia, MMUT-related - **Gene:** MMUT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000255:2-13. **Detection Rate:** Northern European >99%.

Methylmalonic Aciduria and Homocystinuria, cblC Type - **Gene:** MMACHC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015506:1-4. **Detection Rate:** Northern European >99%.

MKS1-related Disorders - **Gene:** MKS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017777:1-18. **Detection Rate:** Northern European >99%.

Mucolipidosis III Gamma - **Gene:** GNPTG. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032520:1-11. **Detection Rate:** Northern European 98%.

Mucolipidosis IV - **Gene:** MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_020533:1-14. **Detection Rate:** Northern European >99%.

Mucopolysaccharidosis Type I - **Gene:** IDUA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000203:1-14. **Detection Rate:** Northern European >99%.

Mucopolysaccharidosis Type II - **Gene:** IDS. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000202:1-9. **Detection Rate:** Northern European 89%.

Mucopolysaccharidosis Type IIIA - **Gene:** SGSH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000199:1-8. **Detection Rate:** Northern European >99%.

Mucopolysaccharidosis Type IIIB - **Gene:** NAGLU. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000263:1-6. **Detection Rate:** Northern European >99%.

Mucopolysaccharidosis Type IIIC - **Gene:** HGSNAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_152419:1-18. **Detection Rate:** Northern European >99%.

Muscular Dystrophy, LAMA2-related - **Gene:** LAMA2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000426:1-43,45-65. **Detection Rate:** Northern European 98%.

MYO7A-related Disorders - **Gene:** MYO7A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000260:2-49. **Detection Rate:** Northern European >99%.

NEB-related Nemaline Myopathy - **Gene:** NEB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001271208:3-80,117-183. **Detection Rate:** Northern European 92%.

Nephrotic Syndrome, NPHS1-related - **Gene:** NPHS1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004646:1-29. **Detection Rate:** Northern European >99%.

Nephrotic Syndrome, NPHS2-related - **Gene:** NPHS2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_014625:1-8. **Detection Rate:** Northern European >99%.

Neuronal Ceroid Lipofuscinosis, CLN6-related - **Gene:** CLN6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017882:1-7. **Detection Rate:** Northern European >99%.

Niemann-Pick Disease Type C1 - **Gene:** NPC1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000271:1-25. **Detection Rate:** Northern European >99%.
Niemann-Pick Disease Type C2 - **Gene:** NPC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_006432:1-5. **Detection Rate:** Northern European >99%.
Niemann-Pick Disease, SMPD1-related - **Gene:** SMPD1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000543:1-6. **Detection Rate:** Northern European >99%.
Nijmegen Breakage Syndrome - **Gene:** NBN. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_002485:1-16. **Detection Rate:** Northern European >99%.
Ornithine Transcarbamylase Deficiency - **Gene:** OTC. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000531:1-10. **Detection Rate:** Northern European 97%.
PCCA-related Propionic Acidemia - **Gene:** PCCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000282:1-24. **Detection Rate:** Northern European 95%.
PCCB-related Propionic Acidemia - **Gene:** PCCB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000532:1-15. **Detection Rate:** Northern European >99%.
PCDH15-related Disorders - **Gene:** PCDH15. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_033056:2-33. **Detection Rate:** Northern European 93%.
Pendred Syndrome - **Gene:** SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000441:2-21. **Detection Rate:** Northern European >99%.
Peroxisome Biogenesis Disorder Type 1 - **Gene:** PEX1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000466:1-24. **Detection Rate:** Northern European >99%.
Peroxisome Biogenesis Disorder Type 3 - **Gene:** PEX12. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000286:1-3. **Detection Rate:** Northern European >99%.
Peroxisome Biogenesis Disorder Type 4 - **Gene:** PEX6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000287:1-17. **Detection Rate:** Northern European 97%.
Peroxisome Biogenesis Disorder Type 5 - **Gene:** PEX2. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000318:4. **Detection Rate:** Northern European >99%.
Peroxisome Biogenesis Disorder Type 6 - **Gene:** PEX10. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153818:1-6. **Detection Rate:** Northern European >99%.
Phenylalanine Hydroxylase Deficiency - **Gene:** PAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000277:1-13. **Detection Rate:** Northern European >99%.
POMGNT-related Disorders - **Gene:** POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017739:2-22. **Detection Rate:** Northern European 96%.
Pompe Disease - **Gene:** GAA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000152:2-20. **Detection Rate:** Northern European 98%.
PPT1-related Neuronal Ceroid Lipofuscinosis - **Gene:** PPT1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000310:1-9. **Detection Rate:** Northern European >99%.
Primary Carnitine Deficiency - **Gene:** SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003060:1-10. **Detection Rate:** Northern European >99%.
Primary Hyperoxaluria Type 1 - **Gene:** AGXT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000030:1-11. **Detection Rate:** Northern European >99%.
Primary Hyperoxaluria Type 2 - **Gene:** GRHPR. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012203:1-9. **Detection Rate:** Northern European >99%.
Primary Hyperoxaluria Type 3 - **Gene:** HOGA1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_138413:1-7. **Detection Rate:** Northern European >99%.
Pycnodysostosis - **Gene:** CTSK. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000396:2-8. **Detection Rate:** Northern European >99%.
Pyruvate Carboxylase Deficiency - **Gene:** PC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000920:3-22. **Detection Rate:** Northern European >99%.
Rhizomelic Chondrodysplasia Punctata Type 1 - **Gene:** PEX7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000288:1-10. **Detection Rate:** Northern European >99%.
RTEL1-related Disorders - **Gene:** RTEL1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_032957:2-35. **Detection Rate:** Northern European >99%.
Sandhoff Disease - **Gene:** HEXB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000521:1-14. **Detection Rate:** Northern European 98%.
Short-chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000017:1-10. **Detection Rate:** Northern European >99%.
Sjogren-Larsson Syndrome - **Gene:** ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000382:1-10. **Detection Rate:** Northern European 96%.
SLC26A2-related Disorders - **Gene:** SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000112:2-3. **Detection Rate:** Northern European >99%.
Smith-Lemli-Opitz Syndrome - **Gene:** DHCR7. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001360:3-9. **Detection Rate:** Northern European >99%.
Spastic Paraplegia Type 15 - **Gene:** ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_015346:2-42. **Detection Rate:** Northern European >99%.
Spinal Muscular Atrophy - **Gene:** SMN1. Autosomal Recessive. Spinal muscular atrophy. **Variant (1):** SMN1 copy number. **Detection Rate:** Northern European 95%.
Spondylothoracic Dysostosis - **Gene:** MESP2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_001039958:1-2. **Detection Rate:** Northern European >99%.
TGM1-related Autosomal Recessive Congenital Ichthyosis - **Gene:** TGM1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000359 2-15. **Detection Rate:** Northern European >99%.
TPP1-related Neuronal Ceroid Lipofuscinosis - **Gene:** TPP1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000391:1-13. **Detection Rate:** Northern European >99%.
Tyrosine Hydroxylase Deficiency - **Gene:** TH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_199292:1-14. **Detection Rate:** Northern European >99%.
Tyrosinemia Type I - **Gene:** FAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000137:1-14. **Detection Rate:** Northern European >99%.
Tyrosinemia Type II - **Gene:** TAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000353:2-12. **Detection Rate:** Northern European >99%.
USH1C-related Disorders - **Gene:** USH1C. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005709:1-21. **Detection Rate:** Northern European >99%.
USH2A-related Disorders - **Gene:** USH2A. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_206933:2-72. **Detection Rate:** Northern European 98%.
Usher Syndrome Type 3 - **Gene:** CLRN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_174878:1-3. **Detection Rate:** Northern European >99%.

Very-long-chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADVL. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000018:1-20. **Detection Rate:** Northern European >99%.
Wilson Disease - **Gene:** ATP7B. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000053:1-21. **Detection Rate:** Northern European >99%.
X-linked Adrenal Hypoplasia Congenita - **Gene:** NR0B1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000475:1-2. **Detection Rate:** Northern European 97%.
X-linked Adrenoleukodystrophy - **Gene:** ABCD1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000033:1-6. **Detection Rate:** Northern European 77%.
X-linked Alport Syndrome - **Gene:** COL4A5. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000495:1-51. **Detection Rate:** Northern European 96%.
X-linked Juvenile Retinoschisis - **Gene:** RS1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000330:1-6. **Detection Rate:** Northern European 98%.
X-linked Myotubular Myopathy - **Gene:** MTM1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000252:2-15. **Detection Rate:** Northern European 96%.
X-linked Severe Combined Immunodeficiency - **Gene:** IL2RG. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000206:1-8. **Detection Rate:** Northern European >99%.
Xeroderma Pigmentosum Group A - **Gene:** XPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000380:1-6. **Detection Rate:** Northern European >99%.
Xeroderma Pigmentosum Group C - **Gene:** XPC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004628:1-16. **Detection Rate:** Northern European 97%.

Risk Calculations

Below are the risk calculations for all conditions tested. Negative results do not rule out the possibility of being a carrier. Residual risk is an estimate of each patient's post-test likelihood of being a carrier, while the reproductive risk represents an estimated likelihood that the patients' future children could inherit each disease. These risks are inherent to all carrier-screening tests, may vary by ethnicity, are predicated on a negative family history, and are present even given a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. In addition, average carrier rates are estimated using incidence or prevalence data from published scientific literature and/or reputable databases, where available, and are incorporated into residual risk calculations for each population/ethnicity. When population-specific data is not available for a condition, average worldwide incidence or prevalence is used. Further, incidence and prevalence data are only collected for the specified phenotypes (which include primarily the classic or severe forms of disease) and may not include alternate or milder disease manifestations associated with the gene. Actual incidence rates, prevalence rates, and carrier rates, and therefore actual residual risks, may be higher or lower than the estimates provided. Carrier rates, incidence/prevalence, and/or residual risks are not provided for some genes with biological or heritable properties that would make these estimates inaccurate. A '†' symbol indicates a positive result. See the full clinical report for interpretation and details. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Disease	DONOR 10622 Residual Risk	Reproductive Risk
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia, HBA1/HBA2-related	Alpha globin status: aa/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	< 1 in 50,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 1,000,000
Argininemia	1 in 12,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 15,000	< 1 in 1,000,000
Aspartylglucosaminuria	< 1 in 50,000	< 1 in 1,000,000
Ataxia with Vitamin E Deficiency	< 1 in 50,000	< 1 in 1,000,000
Ataxia-telangiectasia	1 in 4,200	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 250,000
Autoimmune Polyglandular Syndrome Type 1	1 in 15,000	< 1 in 1,000,000
Autosomal Recessive Osteopetrosis Type 1	1 in 8,900	< 1 in 1,000,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related	1 in 8,100	< 1 in 1,000,000
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	< 1 in 44,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 32,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 42,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	< 1 in 50,000	< 1 in 1,000,000
BCS1L-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Beta-sarcoglycanopathy	1 in 39,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 13,000	1 in 650,000
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 13,000	< 1 in 1,000,000
Canavan Disease	1 in 9,700	< 1 in 1,000,000
Carbamoylphosphate Synthetase I Deficiency	< 1 in 57,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 50,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	1 in 25,000	< 1 in 1,000,000
Cartilage-hair Hypoplasia	< 1 in 50,000	< 1 in 1,000,000
Cerebrotendinous Xanthomatosis	1 in 11,000	< 1 in 1,000,000
Citrullinemia Type 1	1 in 14,000	< 1 in 1,000,000
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 8,600	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
CLN8-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 1 in 1,000,000
COL4A3-related Alport Syndrome	1 in 3,400	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 35,000	< 1 in 1,000,000
Combined Pituitary Hormone Deficiency, PROP1-related	1 in 6,100	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP11B1-related	1 in 8,400	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP21A2-related	1 in 1,300	1 in 280,000
Congenital Disorder of Glycosylation Type Ia	1 in 16,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation, MPI-related	< 1 in 50,000	< 1 in 1,000,000

Disease	DONOR 10622 Residual Risk	Reproductive Risk
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 3,000	1 in 360,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
D-bifunctional Protein Deficiency	1 in 9,000	< 1 in 1,000,000
Delta-sarcoglycanopathy	< 1 in 13,000	< 1 in 1,000,000
Dihydrolipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Dysferlinopathy	1 in 11,000	< 1 in 1,000,000
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)	Not calculated	Not calculated
ERCC6-related Disorders	1 in 8,500	< 1 in 1,000,000
ERCC8-related Disorders	< 1 in 16,000	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,800	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	1 in 9,800	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	1 in 220,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 1,000,000
Familial Hyperinsulinism, ABCC8-related	1 in 17,000	< 1 in 1,000,000
Familial Hyperinsulinism, KCNJ11-related	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	1 in 11,000	< 1 in 1,000,000
Fanconi Anemia Complementation Group A	1 in 2,800	< 1 in 1,000,000
Fanconi Anemia, FANCC-related	< 1 in 50,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 16,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Free Sialic Acid Storage Disorders	< 1 in 30,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 37,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,300	< 1 in 1,000,000
Gaucher Disease	1 in 260	1 in 110,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 2,500	1 in 260,000
GLB1-related Disorders	1 in 17,000	< 1 in 1,000,000
Glutaric Acidemia, GCDH-related	1 in 16,000	< 1 in 1,000,000
Glycine Encephalopathy, AMT-related	1 in 26,000	< 1 in 1,000,000
Glycine Encephalopathy, GLDC-related	1 in 2,500	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 8,700	< 1 in 1,000,000
Glycogen Storage Disease Type Ib	1 in 35,000	< 1 in 1,000,000
Glycogen Storage Disease Type III	1 in 16,000	< 1 in 1,000,000
GNE Myopathy	1 in 23,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 20,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 20,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 3,700	1 in 560,000
Hereditary Fructose Intolerance	1 in 7,900	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 30,000	< 1 in 1,000,000
HMG-CoA Lyase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	< 1 in 1,000,000
Homocystinuria, CBS-related	1 in 9,400	< 1 in 1,000,000
Hydrolethalus Syndrome	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia	1 in 30,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 32,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMB3-related	1 in 32,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 14,000	< 1 in 1,000,000
Leigh Syndrome, French-Canadian Type	< 1 in 50,000	< 1 in 1,000,000
Lipoid Congenital Adrenal Hyperplasia	< 1 in 50,000	< 1 in 1,000,000
Lysosomal Acid Lipase Deficiency	1 in 14,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 39,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ib	1 in 39,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 16,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 4,400	1 in 790,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	< 1 in 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 16,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 1,000,000

Disease	DONOR 10622 Residual Risk	Reproductive Risk
Methylmalonic Acidemia, cblB Type	1 in 48,000	< 1 in 1,000,000
Methylmalonic Acidemia, MMUT-related	1 in 26,000	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 16,000	< 1 in 1,000,000
MKS1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Mucolipidosis III Gamma	< 1 in 20,000	< 1 in 1,000,000
Mucolipidosis IV	< 1 in 50,000	< 1 in 1,000,000
Mucopolysaccharidosis Type I	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type II	< 1 in 1,000,000	1 in 300,000
Mucopolysaccharidosis Type IIIA	1 in 19,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 27,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	< 1 in 50,000	< 1 in 1,000,000
Muscular Dystrophy, LAMA2-related	1 in 5,700	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	1 in 1,200	1 in 400,000
Nephrotic Syndrome, NPHS1-related	< 1 in 50,000	< 1 in 1,000,000
Nephrotic Syndrome, NPHS2-related	1 in 35,000	< 1 in 1,000,000
Neuronal Ceroid Lipofuscinosis, CLN6-related	1 in 20,000	< 1 in 1,000,000
Niemann-Pick Disease Type C1	1 in 19,000	< 1 in 1,000,000
Niemann-Pick Disease Type C2	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-related	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	1 in 16,000	< 1 in 1,000,000
Ornithine Transcarbamylase Deficiency	< 1 in 1,000,000	1 in 140,000
PCCA-related Propionic Acidemia	1 in 4,200	< 1 in 1,000,000
PCCB-related Propionic Acidemia	1 in 22,000	< 1 in 1,000,000
PCDH15-related Disorders	1 in 3,300	< 1 in 1,000,000
Pendred Syndrome	1 in 8,200	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 1	1 in 16,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 3	1 in 44,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 4	1 in 9,300	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 5	< 1 in 71,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 6	< 1 in 50,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	I65T heterozygote †	1 in 200
POMGNT-related Disorders	< 1 in 12,000	< 1 in 1,000,000
Pompe Disease	1 in 4,000	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	1 in 7,700	< 1 in 1,000,000
Primary Carnitine Deficiency	1 in 11,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 17,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	1 in 13,000	< 1 in 1,000,000
Pycnodysostosis	1 in 43,000	< 1 in 1,000,000
Pyruvate Carboxylase Deficiency	1 in 25,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	< 1 in 1,000,000
RTEL1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Sandhoff Disease	1 in 18,000	< 1 in 1,000,000
Short-chain Acyl-CoA Dehydrogenase Deficiency	1 in 11,000	< 1 in 1,000,000
Sjogren-Larsson Syndrome	< 1 in 12,000	< 1 in 1,000,000
SLC26A2-related Disorders	1 in 16,000	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 9,400	< 1 in 1,000,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770	1 in 110,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	< 1 in 1,000,000
Tyrosine Hydroxylase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 16,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 30,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 4,100	< 1 in 1,000,000
Usher Syndrome Type 3	1 in 41,000	< 1 in 1,000,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency	1 in 18,000	< 1 in 1,000,000

Disease	DONOR 10622 Residual Risk	Reproductive Risk
Wilson Disease	1 in 6,500	< 1 in 1,000,000
X-linked Adrenal Hypoplasia Congenita	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 90,000	1 in 42,000
X-linked Alport Syndrome	Not calculated	Not calculated
X-linked Juvenile Retinoschisis	< 1 in 1,000,000	1 in 40,000
X-linked Myotubular Myopathy	Not calculated	Not calculated
X-linked Severe Combined Immunodeficiency	< 1 in 1,000,000	1 in 200,000
Xeroderma Pigmentosum Group A	< 1 in 50,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group C	1 in 7,300	< 1 in 1,000,000