RESULTS RECIPIENT
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Report Date: 02/04/2020

MALE
DONOR 12544
DOB:
Ethnicity: Northern European
Sample Type: EDTA Blood
Date of Collection: 01/28/2020
Date Received: 01/29/2020
Date Tested: 02/04/2020
Barcode: 11004512620213
Accession ID: CSL2DQLKPJZZQNC
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 12544	Partner
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel **(175 conditions tested)**	N/A
POSITIVE: CARRIER Pompe Disease **Reproductive Risk: 1 in 400** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_000152.3(GAA):c.2238G>C (W746C) 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".
POSITIVE: CARRIER Nephrotic Syndrome, NPHS2-related **Reproductive Risk: 1 in 110,000** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_014625.2(NPHS2):c.686G>A(R229Q) 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 8.

CLINICAL NOTES

- None

NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner, as both parents must be carriers before a child is at high risk of developing the disease.
- Genetic counseling is recommended and patients may wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

POSITIVE: CARRIER

Pompe Disease

Reproductive risk: 1 in 400
Risk before testing: 1 in 40,000

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

Patient	**DONOR 12544**	**No partner tested**
Result	✚ Carrier	N/A
Variant(s)	NM_000152.3(GAA):c.2238G>C(W746C) heterozygote	N/A
Methodology	Sequencing with copy number analysis	N/A
Interpretation	This individual is a carrier of Pompe disease. Carriers generally do not experience symptoms.	N/A
Detection rate	98%	N/A
Exons tested	NM_000152:2-20.	N/A

What Is Pompe Disease?

Pompe disease also called glycogen storage disease type II, is an inherited disorder where the body fails to produce enough alpha-glucosidase (also called maltase), an enzyme needed to break down a type of sugar called glycogen. Without adequate amounts of alpha-glucosidase, glycogen builds up in the body, particularly in the muscles, and damages cells. Pompe disease is caused by mutations in the *GAA* gene. People with Pompe disease have muscle weakness that progresses over time, mainly in the muscles used for movement and breathing. The heart may also be affected. The level of alpha-glucosidase remaining is correlated to the severity of symptoms, the age of onset, and disease progression.

Pompe disease is separated into two forms, the infantile-onset form and the late-onset form. These forms are described below.

INFANTILE-ONSET FORM

Infantile-onset Pompe disease is the most severe form because alpha-glucosidase function is entirely absent. Muscle weakness and poor muscle tone causes infants to have trouble moving, holding up their heads, and feeding. They have trouble gaining weight and grow at a slower pace. Infants also have trouble breathing, which can worsen with lung infections. They typically have enlarged hearts, livers, and tongues. Disease progression is usually rapid, and the most common causes of death are heart or lung failure.

LATE-ONSET FORM

Late-onset Pompe disease is less severe because some alpha-glucosidase is still present. Symptoms start with muscle weakness and breathing problems. Some individuals with late-onset Pompe disease have heart problems but without an enlarged heart. They may eventually lose the ability to walk and require a wheelchair, and they may need mechanical assistance to breathe. Disease progression is more gradual, and the most common cause of death is lung failure.

How Common Is Pompe Disease?

The incidence of Pompe disease is approximately 1 in 100,000. Infantile-onset Pompe disease is the most common form.

How Is Pompe Disease Treated?

The FDA has approved enzyme replacement therapy for both infantile-onset and late-onset Pompe disease. Enzyme replacement therapy can help maintain a healthy heart size and normal heart function and may also help improve muscle tone and strength. Individuals need to follow a protein-rich diet, attend physical therapy, and monitor and treat lung infections.

What Is the Prognosis for a Person with Pompe Disease?

In infantile-onset Pompe disease, symptoms may begin at birth but more often begin in the first few months of life. Patients typically die within the first year of life, although enzyme replacement therapy can now prolong life into early childhood. In late-onset Pompe disease, symptoms can begin at any age from childhood to adulthood, and the lifespan depends on how early symptoms begin. The most common cause of death in individuals with Pompe disease is lung failure.

POSITIVE: CARRIER

Nephrotic Syndrome, NPHS2-related

Reproductive risk: 1 in 110,000
Risk before testing: 1 in 310,000

Gene: NPHS2 | **Inheritance Pattern:** Autosomal Recessive

Patient	**DONOR 12544**	**No partner tested**
Result	✚ Carrier	N/A
Variant(s)	NM_014625.2(NPHS2):c.686G>A(R229Q) heterozygote	N/A
Methodology	Sequencing with copy number analysis	N/A
Interpretation	This individual is a carrier of nephrotic syndrome, NPHS2-related. Carriers generally do not experience symptoms. The pathogenicity of R229Q is dependent on the variant observed on the other chromosome.	N/A
Detection rate	>99%	N/A
Exons tested	NM_014625:1-8.	N/A

What Is Nephrotic Syndrome, NPHS2-Related?

Nephrotic syndrome, NPHS2-related is an inherited condition that causes issues with kidney function often leading to kidney failure. Mutations in the *NPHS2* gene cause a form of nephrotic syndrome that is unresponsive to steroid treatment known as steroid-resistant nephrotic syndrome (SRNS). Symptoms of the condition typically begin between 4 and 12 months of age, but in some cases occur later in childhood.

Symptoms of the condition include an excess of protein in the urine (proteinuria), low levels of protein in the blood, kidney failure, and swelling of the body (edema). The swelling can also cause weight gain and high blood pressure. Individuals with nephrotic syndrome are prone to infection due to their inability to retain sufficient amounts of serum antibodies. They are also prone to develop harmful blood clots. Kidney failure typically occurs before the age of 20, and kidney transplantation may allow for a more normal lifespan.

How Common Is Nephrotic Syndrome, NPHS2-Related?

The incidence of all childhood nephrotic syndrome is 2 to 16 per 100,000 individuals worldwide of which 10-20% have SRNS. Approximately 10% of individuals with SRNS carry mutations in the *NPHS2* gene.

How Is Nephrotic Syndrome, NPHS2-Related Treated?

The goal of treatment is to minimize damage to the kidneys. Medication to control blood pressure and high cholesterol may be prescribed. Often children with nephrotic syndrome with protein loss require antibiotics to control for infection. A physician may recommend infusions of protein for children with SRNS to help replace what is lost in the urine. Diuretic drugs may help eliminate excess water and thus reduce swelling while blood thinners may be required to aid in blood clotting. Typically, kidney failure will occur, and a kidney transplant will be required though symptoms of the disease can recur after transplant.

What Is the Prognosis for Nephrotic Syndrome, NPHS2-Related?

The prognosis for an individual with nephrotic syndrome, NPHS2-related varies, but with transplantation and careful medical management, affected children can live into adulthood.

Methods and Limitations

DONOR 12544 [Foresight Carrier Screen]: Sequencing with copy number analysis, spinal muscular atrophy, and analysis of homologous regions.

Sequencing with copy number analysis

High-throughput sequencing and read depth-based copy number analysis are used to analyze the listed exons, as well as selected intergenic and intronic regions, of the genes in the Conditions Tested section of the report. The region of interest (ROI) of the test comprises these regions, in addition to the 20 intronic bases flanking each exon. 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 high coverage and the sequences are compared to standards and references of normal variation. More than 99% of all bases in the ROI are sequenced at greater than the minimum read depth. Mutations may not be detected in areas of lower sequence coverage. Small insertions and deletions may not be as accurately determined as single nucleotide variants. Genes that have closely related pseudogenes may be addressed by a different method. *CFTR* and *DMD* testing includes analysis for both large (exon-level) deletions and duplications with an average sensitivity of 99%, while other genes are only analyzed for large deletions with a sensitivity of >75%. However, the sensitivity may be higher for selected founder deletions. The breakpoints of copy number variants and exons affected are estimated from probe positions. Only exons known to be included in the copy number variant are provided in the name. In some cases, the copy number variant may be larger or smaller than indicated. If *GJB2* is tested, two large upstream deletions which overlap *GJB6* and affect the expression of *GJB2*, del(*GJB6*-D13S1830) and del(*GJB6*-D13S1854), are also analyzed. Mosaicism or somatic variants present at low levels may not be detected. If detected, these may not be reported.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect. Detection rates only account for analytical sensitivity and 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 mutations.

All variants that are a recognized cause of the disease will be reported. In addition, variants that have not previously been established as a recognized cause of disease may be identified. In these cases, only variants classified as "likely" pathogenic are reported. Likely pathogenic variants are described elsewhere in the report as "likely to have a negative impact on gene function". Likely pathogenic variants are evaluated and classified by assessing the nature of the variant and reviewing reports of allele frequencies in cases and controls, functional studies, variant annotation and effect prediction, and segregation studies. Exon level duplications are assumed to be in tandem and are classified according to their predicted effect on the reading frame. Benign variants, variants of uncertain significance, and variants not directly associated with the intended disease phenotype are not reported. Curation summaries of reported variants are available upon request.

Spinal muscular atrophy

Targeted copy number analysis is used to determine the copy number of exon 7 of the *SMN1* gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of *SMN1* are carriers with two *SMN1* genes on one chromosome and a *SMN1* deletion on the other chromosome. This is more likely in individuals who have 2 copies of the *SMN1* gene and are positive for the g.27134T>G SNP, 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 2 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 mutations in certain genes that have homology to other regions. The precise breakpoints of large deletions in these genes cannot be determined, but are estimated from copy number analysis. High numbers of pseudogene copies may interfere with this analysis.

If *CYP21A2* is tested, patients who have one or more additional copies of the *CYP21A2* gene and a loss of function mutation may not actually be a carrier of 21-hydroxylase-deficient congenital adrenal hyperplasia (CAH). Because the true incidence of non-classic CAH is unknown, the residual carrier and reproductive risk numbers on the report are only based on published incidences 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 of overall chances for 21-hydroxylase-deficient CAH, especially in the aforementioned populations, as they do not account for non-classic CAH. If *HBA1*/*HBA2* are tested, some individuals with four alpha globin genes may be carriers, with three genes on one chromosome and a deletion on the other chromosome. This and similar, but rare, carrier states, where complementary changes exist in both the gene and a pseudogene, may not be detected by the assay.

Limitations

In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. This test is designed to detect and report germline alterations. While somatic variants present at low levels may be detected, these may not be reported. f 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. The test does not fully address all inherited forms of intellectual disability, birth defects and genetic disease. A family history of any of these conditions may warrant additional evaluation. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. For example, individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies, which can be identified by CBC and hemoglobin electrophoresis or HPLC (*ACOG Practice Bulletin No. 78. Obstet.Gynecol. 2007;109:229-37*).

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**.

Resources

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

Patients can share their reports via research registries such as Genome Connect, an online research registry working to build the knowledge base about genetics and health. 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

Jack Ji, PhD, FACMG

Report content approved by Jack Ji, PhD, FACMG on Feb 5, 2020

Conditions Tested

11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia - **Gene:** CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000497:1-9. **Detection Rate:** Northern European 94%.

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

ABCC8-related Familial Hyperinsulinism - **Gene:** ABCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000352:1-39. **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 >99%.

Alpha Thalassemia - **Genes:** HBA1, HBA2. Autosomal Recessive. Analysis of homologous regions. **Variants (13):** -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, anti3.7, anti4.2, del HS-40. **Detection Rate:** Unknown due to rarity of disease.

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%.

AMT-related Glycine Encephalopathy - **Gene:** AMT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000481:1-9. **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 98%.

ATP7A-related Disorders - **Gene:** ATP7A. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000052:2-23. **Detection Rate:** Northern European 96%.

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 >99%.

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%.

CLN6-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_017882:1-7. **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 97%.
COL4A4-related Alport Syndrome - **Gene:** COL4A4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000092:2-48. **Detection Rate:** Northern European 98%.
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, 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, V281L, [(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 99%.
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 99%.
ERCC8-related Disorders - **Gene:** ERCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000082:1-12. **Detection Rate:** Northern European 95%.
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 >99%.
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:** IKBKAP. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_003640:2-37. **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%.
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 88%.
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%.
GLDC-related Glycine Encephalopathy - **Gene:** GLDC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000170:1-25. **Detection Rate:** Northern European 94%.
Glutaric Acidemia, GCDH-related - **Gene:** GCDH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000159:2-12. **Detection Rate:** Northern European >99%.
Glycogen Storage Disease Type Ia - **Gene:** G6PC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000151:1-5. **Detection Rate:** Northern European >99%.
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%.
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related - **Gene:** LAMB3. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000228 2-23. **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 98%.
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, LAMC2-related - **Gene:** LAMC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_005562:1-23. **Detection Rate:** Northern European >99%.
KCNJ11-related Familial Hyperinsulinism - **Gene:** KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. **Exon:** NM_000525:1. **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%.
LAMA2-related Muscular Dystrophy - **Gene:** LAMA2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000426:1-65. **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 >99%.
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 96%.
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 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 >99%.
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 88%.
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%.
MUT-related Methylmalonic Acidemia - **Gene:** MUT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000255:2-13. **Detection Rate:** Northern European >99%.
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%.
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%.
Salla Disease - **Gene:** SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_012434:1-11. **Detection Rate:** Northern European 98%.
Sandhoff Disease - **Gene:** HEXB. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000521:1-14. **Detection Rate:** Northern European >99%.
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 94%.
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 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 95%.
X-linked Congenital Adrenal Hypoplasia - **Gene:** NR0B1. X-linked Recessive. Sequencing with copy number analysis. **Exons:** NM_000475:1-2. **Detection Rate:** Northern European 99%.
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 98%.
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. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's 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 after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

†Indicates a positive result. See the full clinical report for interpretation and details.

Disease	DONOR 12544 Residual Risk	Reproductive Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 3,800	< 1 in 1,000,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 1,000,000
ABCC8-related Familial Hyperinsulinism	1 in 17,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia	Alpha globin status: aa/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	1 in 45,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 1,000,000
AMT-related Glycine Encephalopathy	1 in 22,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 1,000,000
Argininemia	< 1 in 17,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 13,000	< 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 11,000	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 600,000
Autoimmune Polyglandular Syndrome Type 1	1 in 15,000	< 1 in 1,000,000
Autosomal Recessive Osteopetrosis Type 1	1 in 35,000	< 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
CLN6-related Neuronal Ceroid Lipofuscinosis	1 in 43,000	< 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 6,200	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 12,000	< 1 in 1,000,000
Combined Pituitary Hormone Deficiency, PROP1-related	1 in 6,100	< 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
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 1,000,000

Disease	DONOR 12544 Residual Risk	Reproductive Risk
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 40,000	< 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 26,000	< 1 in 1,000,000
ERCC8-related Disorders	< 1 in 9,900	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,500	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	< 1 in 50,000	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	1 in 80,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	< 1 in 50,000	< 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
Galactokinase Deficiency	1 in 10,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,000	< 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 19,000	< 1 in 1,000,000
GLDC-related Glycine Encephalopathy	1 in 2,800	< 1 in 1,000,000
Glutaric Acidemia, GCDH-related	1 in 16,000	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 18,000	< 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 32,000	< 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 3,100	1 in 390,000
Hereditary Fructose Intolerance	1 in 7,900	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related	< 1 in 50,000	< 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 33,000	< 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 27,000	< 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, LAMC2-related	< 1 in 50,000	< 1 in 1,000,000
KCNJ11-related Familial Hyperinsulinism	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 14,000	< 1 in 1,000,000
LAMA2-related Muscular Dystrophy	1 in 34,000	< 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 18,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 42,000	< 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 13,000	< 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
Methylmalonic Acidemia, cblB Type	1 in 48,000	< 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

Disease	DONOR 12544 Residual Risk	Reproductive Risk
Mucolipidosis III Gamma	< 1 in 50,000	< 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 600,000	1 in 150,000
Mucopolysaccharidosis Type IIIA	1 in 12,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 25,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	1 in 37,000	< 1 in 1,000,000
MUT-related Methylmalonic Acidemia	1 in 26,000	< 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	NM_014625.2(NPHS2):c.686G>A(R229Q) heterozygote †	1 in 110,000
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	1 in 4,800	1 in 940,000
POMGNT-related Disorders	< 1 in 12,000	< 1 in 1,000,000
Pompe Disease	NM_000152.3(GAA):c.2238G>C(W746C) heterozygote †	1 in 400
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
Salla Disease	< 1 in 30,000	< 1 in 1,000,000
Sandhoff Disease	1 in 32,000	< 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 35,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 2,200	< 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
Wilson Disease	1 in 8,600	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 90,000	1 in 42,000
X-linked Alport Syndrome	Not calculated	Not calculated

Disease	DONOR 12544 Residual Risk	Reproductive Risk
X-linked Congenital Adrenal Hypoplasia	< 1 in 1,000,000	< 1 in 1,000,000
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