Counsyl

RESULTS RECIPIENT
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Report Date: 06/28/2017

FEMALE
SHANTEL MACKIE
DOB: 04/18/1973
Ethnicity: Northern European
Sample Type: OG-510 Saliva
Date of Collection: 06/23/2017
Date Received: 06/28/2017
Date Tested: 06/28/2017
Barcode: 55200000016169
Indication: Screening for genetic disease carrier status

MALE
HIRAM MONK
DOB: 05/29/1989
Ethnicity: Northern European
Sample Type: OG-510 Saliva
Date of Collection: 06/23/2017
Date Received: 06/28/2017
Date Tested: 06/28/2017
Barcode: 55200000016492
Indication: Screening for genetic disease carrier status

Foresight™ Carrier Screen

POSITIVE: HIGH REPRODUCTIVE RISK

ABOUT THIS TEST

The **Counsyl 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	SHANTEL MACKIE	HIRAM MONK
Panel Information	Foresight Carrier Screen Universal Panel with Fragile X Syndrome **(176 conditions tested)**	Foresight Carrier Screen Universal Panel **(175 conditions tested)**
POSITIVE: HIGH REPRODUCTIVE RISK Smith-Lemli-Opitz Syndrome **Reproductive Risk: 1 in 4** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_001360.2(DHCR7):c.964-1G>C (aka IVS8-1G>C) heterozygote	**CARRIER*** NM_001360.2(DHCR7):c.964-1G>C (aka IVS8-1G>C) heterozygote

*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 6.

CLINICAL NOTES

- None

NEXT STEPS

- 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.

Counsyl has renamed its products effective TBD. The Family Prep Screen is now the Foresight Carrier Screen. The new names now appear on all communications from Counsyl. If you have any questions, please contact Counsyl directly.

POSITIVE: HIGH REPRODUCTIVE RISK

Smith-Lemli-Opitz Syndrome

Reproductive risk: 1 in 4
Risk before testing: 1 in 9,800

Gene: DHCR7 | **Inheritance Pattern:** Autosomal Recessive

Patient	SHANTEL MACKIE	HIRAM MONK
Result	⊕ Carrier	⊕ Carrier
Variant(s)	NM_001360.2(DHCR7):c.964-1G>C(aka IVS8-1G>C) heterozygote	NM_001360.2(DHCR7):c.964-1G>C(aka IVS8-1G>C) heterozygote
Methodology	Sequencing with copy number analysis	Sequencing with copy number analysis
Interpretation	This individual is a carrier of Smith-Lemli-Opitz syndrome. Carriers generally do not experience symptoms. The c.964-1G>C mutation is associated with the severe form of this disease.	This individual is a carrier of Smith-Lemli-Opitz syndrome. Carriers generally do not experience symptoms. The c.964-1G>C mutation is associated with the severe form of this disease.
Detection rate	>99%	>99%
Exons tested	NM_001360:3-9.	NM_001360:3-9.

What is Smith-Lemli-Opitz Syndrome?

Smith-Lemli-Opitz syndrome, or SLO syndrome, is an inherited disorder in which the body's ability to make cholesterol is impaired due to a deficient enzyme. Cholesterol is critical for the structure of cells, and is necessary for normal fetal development. It also plays an important role in the production of hormones and digestive acids. In addition to low cholesterol levels, SLO syndrome also causes toxic byproducts of cholesterol production to build up throughout the body, further disrupting growth and development.

In children with little or no ability to make cholesterol, symptoms are severe. These infants are commonly born with an abnormally small head, cleft palate, and weak muscle tone. They often have difficulty feeding because they lack the sucking reflex or have an abnormally small stomach that causes persistent vomiting. Some have extra fingers or toes as well as the typical fused second and third toes on both feet. Male infants may have deformed or underdeveloped genitalia.

Infants with the severe form of SLO syndrome grow slowly and 90% have moderate to severe mental disability. Severely affected infants may also have heart defects and problems with their kidneys, causing death in the first months of life.

Some children are born with a milder form of the condition in which the body can produce some cholesterol. Symptoms may include developmental delays, feet with the second and third toes fused together, slow growth, and short stature. These children generally learn to walk and talk and can acquire other skills, although they can rarely live independently as adults. Adults with the disease often show aggressive behavior.

Symptoms of the disease can vary from person to person. Some affected people have only minor symptoms of the condition.

How common is Smith-Lemli-Opitz Syndrome?

Smith-Lemli-Opitz syndrome affects an estimated 1 in 20,000 to 60,000 people. This disease is more common in those of European ancestry, particularly those in Slovakia and the Czech Republic. It is very rare among people of African and Asian descent.

How is Smith-Lemli-Opitz Syndrome treated?

There is no cure for SLO syndrome, but its symptoms can be addressed. The primary treatment is to supplement the person's diet with large amounts of dietary cholesterol, either in the form of purified cholesterol or in foods such as egg yolks and cream. This has been shown to improve symptoms. Early intervention and therapy helps with speech and physical disabilities. Medication may treat symptoms such as vomiting, constipation, and gastroesophageal reflux. Surgery and orthotics can help muscle spasms and improve mobility.

Because the condition can cause extreme sun sensitivity, people with SLO syndrome should always wear sunblock, sunglasses, and appropriate clothing when they go outdoors.

What is the prognosis for a person with Smith-Lemli-Opitz Syndrome?

Although serious internal malformations can lead to early death, with good nutrition and medical care many people with SLO syndrome can have a normal lifespan. Mental disability typically prevents people with this disease from living independently.

Methods and Limitations

SHANTEL MACKIE [Foresight Carrier Screen]: sequencing with copy number analysis, triplet repeat detection, spinal muscular atrophy, and analysis of homologous regions.
HIRAM MONK [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. 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.

Triplet repeat detection

PCR is used to size the CGG repeat in the 5' UTR of *FMR1* (NM_002024.4: c.1-131CGG[1_n]). PCR products generated from fluorescently labeled primers are detected by capillary electrophoresis. Reported sizes are accurate to +/- 1 repeat for up to 200 repeats. Alleles above 200 CGG repeats (full mutations), while identified, are not sized. Nearby mutations may interfere with detection of CGG repeat expansions. Deletion of the CGG repeat region and other similar *FMR1* mutations may not be detectable. Methylation is not analyzed. Small degrees of size mosaicism, including gonadal mosaicism, may not be detected as the test has been calibrated to yield results that are equivalent to the results from Southern blot.

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.

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. 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. 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 Counsyl, 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**.

LAB DIRECTORS

H. Peter Kang, MD, MS, FCAP

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%.

21-hydroxylase-deficient Congenital Adrenal Hyperplasia - **Gene:** CYP21A2. Autosomal Recessive. Analysis of Homologous Regions. **Variants (13):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111VfsX21, I173N, L308FfsX6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Northern European 96%.

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 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/--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_001244438:1-8. **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%.

ARSACS - **Gene:** SACS. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_014363:2-10. **Detection Rate:** Northern European 99%.

Aspartylglycosaminuria - **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%.

Autosomal Recessive Osteopetrosis Type 1 - **Gene:** TCIRG1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_006019:2-20. **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%.

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%.

Cohen Syndrome - **Gene:** VPS13B. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_017890:2-62. **Detection Rate:** Northern European 96%.

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%.

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 Ib - **Gene:** MPI. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_002435:1-8. **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 Finnish Nephrosis - **Gene:** NPHS1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_004646:1-29. **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%.

Dysferlinopathy - **Gene:** DYSF. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001130987:1-56. **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 Type C - **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%.

Fragile X Syndrome - **Gene:** FMR1. X-linked Dominant. Triplet Repeat Detection. **Variant (1):** FMR1 CGG repeat number. **Detection Rate:** Northern European >99%.
Hiram (Male): Not tested.

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):** D448H, D448V, L483P, N409S, R502C, R502H, R535H, V433L, c.115+1G>A, c.84dupG. **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 Type 1 - **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%.

GNPTAB-related Disorders - **Gene:** GNPTAB. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_024312:1-21. **Detection Rate:** Northern European >99%.

GRACILE Syndrome - **Gene:** BCS1L. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_004328:3-9. **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, LAMA3-related - **Gene:** LAMA3. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000227:1-38. **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%.

Herlitz Junctional Epidermolysis Bullosa, LAMC2-related - **Gene:** LAMC2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_005562:1-23. **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 Caused by Cystathionine Beta-synthase Deficiency - **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_001134793:3. **Detection Rate:** Northern European >99%.

Hypophosphatasia, Autosomal Recessive - **Gene:** ALPL. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000478:2-12. **Detection Rate:** Northern European >99%.

Inclusion Body Myopathy 2 - **Gene:** GNE. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001128227:1-12. **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%.
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%.
Lipoamide Dehydrogenase Deficiency - **Gene:** DLD. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000108:1-14. **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 1B - **Gene:** BCKDHB. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_183050:1-10. **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 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%.
Muscle-eye-brain Disease - **Gene:** POMGNT1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_017739:2-22. **Detection Rate:** Northern European 96%.
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%.
Niemann-Pick Disease Type C - **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-associated - **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%.
Northern Epilepsy - **Gene:** CLN8. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_018941:2-3. **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_001178014:1-16. **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 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%.
PEX1-related Zellweger Syndrome Spectrum - **Gene:** PEX1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000466:1-24. **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%.
PKHD1-related Autosomal Recessive Polycystic Kidney Disease - **Gene:** PKHD1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_138694:2-67. **Detection Rate:** Northern European >99%.

Polyglandular Autoimmune Syndrome Type 1 - **Gene:** AIRE. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000383:1-14. **Detection Rate:** Northern European >99%.
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%.
PROP1-related Combined Pituitary Hormone Deficiency - **Gene:** PROP1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_006261:1-3. **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_022172:2-21. **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%.
Segawa Syndrome - **Gene:** TH. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000360:1-13. **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 97%.
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%.
Steroid-resistant Nephrotic Syndrome - **Gene:** NPHS2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_014625:1-8. **Detection Rate:** Northern European >99%.
Sulfate Transporter-related Osteochondrodysplasia - **Gene:** SLC26A2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000112:2-3. **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%.
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_153676:1-27. **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 each patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood 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 after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation.

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

Disease	SHANTEL MACKIE Residual Risk	HIRAM MONK Residual Risk	Reproductive Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 3,800	1 in 3,800	< 1 in 1,000,000
21-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 1,400	1 in 1,400	< 1 in 1,000,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
ABCC8-related Hyperinsulinism	1 in 11,000	1 in 11,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia	Alpha globin status: aa/aa.	Alpha globin status: aa/aa.	Low
Alpha-mannosidosis	1 in 35,000	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	1 in 45,000	1 in 45,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
AMT-related Glycine Encephalopathy	1 in 22,000	1 in 22,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Argininemia	< 1 in 17,000	< 1 in 17,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 13,000	1 in 13,000	< 1 in 1,000,000
ARSACS	< 1 in 44,000	< 1 in 44,000	< 1 in 1,000,000
Aspartylglycosaminuria	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Ataxia with Vitamin E Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Ataxia-telangiectasia	1 in 8,200	1 in 8,200	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	< 1 in 1,000,000	< 1 in 1,000,000
Autosomal Recessive Osteopetrosis Type 1	1 in 35,000	1 in 35,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Beta-sarcoglycanopathy	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 12,000	1 in 12,000	< 1 in 1,000,000
Bloom Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 13,000	1 in 13,000	< 1 in 1,000,000
Canavan Disease	< 1 in 31,000	< 1 in 31,000	< 1 in 1,000,000
Carbamoylphosphate Synthetase I Deficiency	< 1 in 57,000	< 1 in 57,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Cartilage-hair Hypoplasia	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Cerebrotendinous Xanthomatosis	1 in 11,000	1 in 11,000	< 1 in 1,000,000
Citrullinemia Type 1	1 in 12,000	1 in 12,000	< 1 in 1,000,000
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 22,000	1 in 22,000	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
CLN6-related Neuronal Ceroid Lipofuscinosis	1 in 43,000	1 in 43,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 12,000	< 1 in 12,000	< 1 in 1,000,000
COL4A3-related Alport Syndrome	1 in 6,200	1 in 6,200	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 12,000	1 in 12,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ia	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ib	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Congenital Finnish Nephrosis	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 2,700	1 in 2,700	< 1 in 1,000,000
Cystinosis	1 in 22,000	1 in 22,000	< 1 in 1,000,000
D-bifunctional Protein Deficiency	1 in 9,000	1 in 9,000	< 1 in 1,000,000
Delta-sarcoglycanopathy	< 1 in 40,000	< 1 in 40,000	< 1 in 1,000,000

Disease	SHANTEL MACKIE Residual Risk	HIRAM MONK Residual Risk	Reproductive Risk
Dysferlinopathy	1 in 11,000	1 in 11,000	< 1 in 1,000,000
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)	Not calculated	Not calculated	Not calculated
ERCC6-related Disorders	1 in 26,000	1 in 26,000	< 1 in 1,000,000
ERCC8-related Disorders	< 1 in 9,900	< 1 in 9,900	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,500	1 in 7,500	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	< 1 in 1,000,000	< 1 in 1,000,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Fanconi Anemia Complementation Group A	1 in 2,800	1 in 2,800	< 1 in 1,000,000
Fanconi Anemia Type C	1 in 16,000	1 in 16,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 16,000	1 in 16,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Fragile X Syndrome	Normal: 29 and 31 repeats	Not tested	Not calculated
Galactokinase Deficiency	1 in 10,000	1 in 10,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	1 in 8,600	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,000	1 in 3,000	< 1 in 1,000,000
Gaucher Disease	1 in 280	1 in 280	1 in 310,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 3,200	1 in 3,200	< 1 in 1,000,000
GLB1-related Disorders	1 in 19,000	1 in 19,000	< 1 in 1,000,000
GLDC-related Glycine Encephalopathy	1 in 2,800	1 in 2,800	< 1 in 1,000,000
Glutaric Acidemia Type 1	1 in 10,000	1 in 10,000	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 18,000	1 in 18,000	< 1 in 1,000,000
Glycogen Storage Disease Type Ib	1 in 35,000	1 in 35,000	< 1 in 1,000,000
Glycogen Storage Disease Type III	1 in 16,000	1 in 16,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 32,000	1 in 32,000	< 1 in 1,000,000
GRACILE Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 15,000	1 in 15,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 5,000	1 in 5,000	< 1 in 1,000,000
Hereditary Fructose Intolerance	1 in 8,000	1 in 8,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 30,000	1 in 30,000	< 1 in 1,000,000
HMG-CoA Lyase Deficiency	< 1 in 33,000	< 1 in 33,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	1 in 15,000	< 1 in 1,000,000
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency	1 in 25,000	1 in 25,000	< 1 in 1,000,000
Hydrolethalus Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia, Autosomal Recessive	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Inclusion Body Myopathy 2	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 25,000	1 in 25,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
KCNJ11-related Familial Hyperinsulinism	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 15,000	1 in 15,000	< 1 in 1,000,000
LAMA2-related Muscular Dystrophy	1 in 34,000	1 in 34,000	< 1 in 1,000,000
Leigh Syndrome, French-Canadian Type	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Lipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Lipoid Congenital Adrenal Hyperplasia	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Lysosomal Acid Lipase Deficiency	1 in 18,000	1 in 18,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type 1B	1 in 25,000	1 in 25,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 42,000	1 in 42,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 13,000	1 in 13,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 5,900	1 in 5,900	< 1 in 1,000,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 20,000	1 in 20,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblB Type	1 in 48,000	1 in 48,000	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 16,000	1 in 16,000	< 1 in 1,000,000
MKS1-related Disorders	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Mucolipidosis III Gamma	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Mucolipidosis IV	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000

Disease	SHANTEL MACKIE Residual Risk	HIRAM MONK Residual Risk	Reproductive Risk
Mucopolysaccharidosis Type I	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type II	1 in 300,000	< 1 in 1,000,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIA	1 in 12,000	1 in 12,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 25,000	1 in 25,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	1 in 37,000	1 in 37,000	< 1 in 1,000,000
Muscle-eye-brain Disease	< 1 in 12,000	< 1 in 12,000	< 1 in 1,000,000
MUT-related Methylmalonic Acidemia	1 in 26,000	1 in 26,000	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	< 1 in 6,700	< 1 in 6,700	< 1 in 1,000,000
Niemann-Pick Disease Type C	1 in 19,000	1 in 19,000	< 1 in 1,000,000
Niemann-Pick Disease Type C2	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-associated	1 in 25,000	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Northern Epilepsy	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Ornithine Transcarbamylase Deficiency	< 1 in 1,000,000	< 1 in 1,000,000	< 1 in 1,000,000
PCCA-related Propionic Acidemia	1 in 4,200	1 in 4,200	< 1 in 1,000,000
PCCB-related Propionic Acidemia	1 in 22,000	1 in 22,000	< 1 in 1,000,000
PCDH15-related Disorders	1 in 5,300	1 in 5,300	< 1 in 1,000,000
Pendred Syndrome	1 in 7,000	1 in 7,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 3	1 in 44,000	1 in 44,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 4	1 in 9,300	1 in 9,300	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 5	< 1 in 71,000	< 1 in 71,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 6	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
PEX1-related Zellweger Syndrome Spectrum	1 in 11,000	1 in 11,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 5,000	1 in 5,000	< 1 in 1,000,000
PKHD1-related Autosomal Recessive Polycystic Kidney Disease	1 in 6,100	1 in 6,100	< 1 in 1,000,000
Polyglandular Autoimmune Syndrome Type 1	1 in 14,000	1 in 14,000	< 1 in 1,000,000
Pompe Disease	1 in 6,300	1 in 6,300	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Primary Carnitine Deficiency	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 35,000	1 in 35,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	1 in 13,000	1 in 13,000	< 1 in 1,000,000
PROP1-related Combined Pituitary Hormone Deficiency	1 in 11,000	1 in 11,000	< 1 in 1,000,000
Pycnodysostosis	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Pyruvate Carboxylase Deficiency	1 in 25,000	1 in 25,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	1 in 16,000	< 1 in 1,000,000
RTEL1-related Disorders	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Salla Disease	< 1 in 30,000	< 1 in 30,000	< 1 in 1,000,000
Sandhoff Disease	1 in 32,000	1 in 32,000	< 1 in 1,000,000
Segawa Syndrome	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Short Chain Acyl-CoA Dehydrogenase Deficiency	1 in 16,000	1 in 16,000	< 1 in 1,000,000
Sjogren-Larsson Syndrome	1 in 9,100	1 in 9,100	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	c.964-1G>C heterozygote †	c.964-1G>C heterozygote †	1 in 4
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 630	SMN1: 2 copies 1 in 630	< 1 in 1,000,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Steroid-resistant Nephrotic Syndrome	1 in 40,000	1 in 40,000	< 1 in 1,000,000
Sulfate Transporter-related Osteochondrodysplasia	1 in 11,000	1 in 11,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	1 in 30,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 17,000	1 in 17,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 35,000	1 in 35,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 2,200	1 in 2,200	< 1 in 1,000,000
Usher Syndrome Type 3	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	1 in 8,800	1 in 8,800	< 1 in 1,000,000
Wilson Disease	1 in 8,600	1 in 8,600	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 45,000	1 in 390,000	1 in 180,000
X-linked Alport Syndrome	Not calculated	Not calculated	Not calculated
X-linked Congenital Adrenal Hypoplasia	< 1 in 1,000,000	< 1 in 1,000,000	< 1 in 1,000,000

RESULTS RECIPIENT
UNIVERSITY MEDICAL CENTER 235
Attn: Dr. Paul Smith
NPI: 0000000006
Report Date: 06/28/2017

FEMALE
SHANTEL MACKIE
DOB: 04/18/1973
Ethnicity: Northern European
Barcode: 55200000016169

MALE
HIRAM MONK
DOB: 05/29/1989
Ethnicity: Northern European
Barcode: 55200000016492

Disease	SHANTEL MACKIE Residual Risk	HIRAM MONK Residual Risk	Reproductive Risk
X-linked Juvenile Retinoschisis	1 in 840,000	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Myotubular Myopathy	Not calculated	Not calculated	Not calculated
X-linked Severe Combined Immunodeficiency	< 1 in 1,000,000	< 1 in 1,000,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group A	< 1 in 50,000	< 1 in 50,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group C	1 in 7,300	1 in 7,300	< 1 in 1,000,000