

RESULTS RECIPIENT

SEATTLE SPERM BANK

Attn: Jeffrey Olliffe 4915 25th Ave NE Ste 204W Seattle, WA 98105

Phone: (206) 588-1484 Fax: (206) 466-4696 NPI: 1306838271 Report Date: 05/27/2021 MALE DONOR 14226

Ethnicity: Northern European Sample Type: EDTA Blood Date of Collection: 05/14/2021 Date Received: 05/17/2021 Date Tested: 05/26/2021

Barcode: Accession ID:

Indication: Egg or sperm donor

FEMALE N/A

POSITIVE: CARRIER

Foresight® Carrier Screen

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 14226	Partner	
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel (175 conditions tested)	N/A	
POSITIVE: CARRIER Sandhoff Disease	CARRIER* NM_000521.3(HEXB):c.1250C>T (P417L) 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".	
Reproductive Risk: 1 in 1,300 Inheritance: Autosomal Recessive	(i 417 L) Neterozygote		

^{*}Carriers generally do not experience symptoms.

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

CLINICAL NOTES

None

NEXT STEPS

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



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Positive: carrier Sandhoff Disease

Gene: HEXB | Inheritance Pattern: Autosomal Recessive

Reproductive risk: 1 in 1,300 Risk before testing: 1 in 400,000

Patient	DONOR 14226	No partner tested
Result	□ Carrier	N/A
Variant(s)	NM_000521.3(HEXB):c.1250C>T(P417L) heterozygote	N/A
Methodology	Sequencing with copy number analysis (v3.1)	N/A
Interpretation	This individual is a carrier of Sandhoff disease. Carriers generally do not experience symptoms.	N/A
Detection rate	98%	N/A
Exons tested	NM_000521:1-14.	N/A

What is Sandhoff Disease?

Sandhoff disease is an inherited, lysosomal storage disorder caused by harmful genetic changes (mutations) in the HEXB gene. The HEXB gene contains instructions for an enzyme called β -hexosaminidase, which is responsible for breaking down harmful substances, primarily a fatty protein known as GM2 ganglioside. Normally, GM2 ganglioside is broken down inside cells and is harmless. In individuals with Sandhoff disease, GM2 ganglioside and other molecules do not get broken down. These molecules then build up in the brain and damage nerve cells. The disease has been broken down into three different types, based on when symptoms first appear.

INFANTILE (CLASSIC) FORM

The classic form of Sandhoff disease appears shortly after birth and is the most severe form of the disease. At around three to six months of age, affected infants start to lose milestones and their muscles will become weaker. They may not be able to roll over, sit up, or crawl. Affected infants will also have an exaggerated startle reaction to noises or touch. Over time, children with Sandhoff disease will develop seizures, vision and hearing loss, intellectual disability, and paralysis. Other symptoms include enlarged organs (organomegaly) and bone abnormalities. A red spot in the eye known as a "cherry-red spot" is characteristic of many lysosomal disorders, including Sandhoff disease.

JUVENILE-ONSET FORM

A milder, rarer form of Sandhoff disease occurs when an individual has mutations that only cause a partial enzyme deficiency. Signs and symptoms vary widely and can begin in childhood or adolescence. Affected individuals may experience muscle weakness, difficulty coordinating movement, speech problems, recurrent respiratory infections, and seizures.

LATE-ONSET FORM

The late-onset form can be difficult to diagnose. Early signs can include clumsiness and muscle weakness in the legs. Over time, people with late-onset Sandhoff disease may require mobility assistance and have difficulty with speech and swallowing. About 40% of affected adults experience mental illness, such as bipolar disorder or psychotic episodes.



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How common is Sandhoff Disease?

The incidence of Sandhoff disease is estimated to be 1 in 400,000. The condition may be observed more frequently in certain populations including the Creole population of northern Argentina, the Metis Indians in Saskatchewan, Canada, and people from Lebanon.

How is Sandhoff Disease treated?

Treatment for Sandhoff disease includes supportive care for symptoms, such as medications to control seizures, assistance in getting adequate nutrition, and breathing assistance. There is no cure for the disease.

What is the prognosis for an individual with Sandhoff Disease?

Children with the severe infantile-onset form will often have recurrent seizures by age two and eventually lose muscle function, mental function, and sight, becoming mostly non-responsive to their environment. Death usually occurs by age three and is generally caused by respiratory infections. Children with juvenile-onset Sandhoff disease will show similar health problems, but at an older age, and will also progressively decline. Though challenging and debilitating, the late-onset form does not always shorten life span.



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Methods and Limitations

DONOR 14226 [Foresight Carrier Screen]: Sequencing with copy number analysis, spinal muscular atrophy, analysis of homologous regions, and alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (Assay(s): DTS v3.2).

Sequencing with copy number analysis

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

Spinal muscular atrophy

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

Analysis of homologous regions

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

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



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

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

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

Interpretation of reported variants

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

Limitations

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

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

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



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Incidental Findings

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

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

Resources

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

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

SENIOR LABORATORY DIRECTOR

Karla R. Bowles, PhD, FACMG, CGMB

Kenle R. Boules

Report content approved by Karla Bowles, PhD, FACMG, CGMB on May 27, 2021



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Conditions Tested

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

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

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

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

Alpha-sarcoglycanopathy - **Gene:** SGCA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000023:1-9. **Detection Rate:** Northern European >90%

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

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

Argininemia - Gene: ARG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000045:1-8. Detection Rate: Northern European 97%. Argininosuccinic Aciduria - Gene: ASL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001024943:1-16. Detection Rate: Northern European >99%.

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

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

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

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

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

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

Autosomal Recessive Polycystic Kidney Disease, PKHD1-related - Gene: PKHD1. Autosomal Recessive. Sequencing with copy number analysis. Exons:

NM_138694 2-67. Detection Rate: Northern European >99%. Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay - Gene: SACS.

Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_014363 2-10. Detection Rate: Northern European 99%.

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

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

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

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

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

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

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

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

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

Canavan Disease - Gene: ASPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000049:1-6. Detection Rate: Northern European 98%. Carbamoylphosphate Synthetase I Deficiency - Gene: CPS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001875:1-38. Detection Rate: Northern European >99%.

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

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

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

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

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

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

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

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

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

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



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>99%.

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Familial Hyperinsulinism, ABCC8-related - Gene: ABCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000352:1-39. Detection Rate: Northern European >99%.

Familial Hyperinsulinism, KCNJ11-related - Gene: KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_000525:1. Detection Rate: Northern European >99%.

Familial Mediterranean Fever - Gene: MEFV. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000243:1-10. Detection Rate: Northern European >99%.

Fanconi Anemia Complementation Group A - Gene: FANCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000135:1-43. Detection Rate: Northern European 92%.

Fanconi Anemia, FANCC-related - Gene: FANCC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000136:2-15. **Detection Rate:** Northern European >99%.

FKRP-related Disorders - Gene: FKRP. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_024301:4. Detection Rate: Northern European >99%. FKTN-related Disorders - Gene: FKTN. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001079802:3-11. Detection Rate: Northern European

Free Sialic Acid Storage Disorders - Gene: SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_012434:1-11. Detection Rate: Northern European 98%.

Galactokinase Deficiency - **Gene**: GALK1. Autosomal Recessive. Sequencing with copy number analysis. **Exons**: NM_000154:1-8. **Detection Rate**: Northern European >99%

Galactosemia - Gene: GALT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000155:1-11. Detection Rate: Northern European >99%.

Gamma-sarcoglycanopathy - Gene: SGCG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000231:2-8. Detection Rate: Northern European 87%.

Gaucher Disease - Gene: GBA. Autosomal Recessive. Analysis of homologous regions. Variants (10): D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs*18. Detection Rate: Northern European 60%.

GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness - Gene: GJB2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004004:1-2. Detection Rate: Northern European >99%.

GLB1-related Disorders - **Gene**: GLB1. Autosomal Recessive. Sequencing with copy number analysis. **Exons**: NM_000404:1-16. **Detection Rate**: Northern European >99%

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%.

Glycine Encephalopathy, AMT-related - Gene: AMT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000481:1-9. **Detection Rate:** Northern European >99%.

Glycogen Storage Disease Type la - **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%.

COL4A4-related Alport Syndrome - Gene: COL4A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000092:2-48. Detection Rate: Northern European >99%.

Combined Pituitary Hormone Deficiency, PROP1-related - Gene: PROP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006261:1-3. Detection Rate: Northern European >99%.

Congenital Adrenal Hyperplasia, CYP11B1-related - Gene: CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000497:1-9. Detection Rate: Northern European 97%.

Congenital Adrenal Hyperplasia, CYP21A2-related - Gene: CYP21A2. Autosomal Recessive. Analysis of homologous regions. Variants (13): CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G. Detection Rate: Northern European 96%.

Congenital Disorder of Glycosylation Type Ia - Gene: PMM2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000303:1-8. Detection Rate: Northern European >99%.

Congenital Disorder of Glycosylation Type Ic - Gene: ALG6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_013339:2-15. Detection Rate: Northern European >99%.

Congenital Disorder of Glycosylation, MPI-related - Gene: MPI. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_002435:1-8. Detection Rate: Northern European >99%.

Costeff Optic Atrophy Syndrome - Gene: OPA3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_025136:1-2. Detection Rate: Northern European >99%.

Cystic Fibrosis - Gene: CFTR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. Detection Rate: Northern European >99%.

Cystinosis - **Gene:** CTNS. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_004937:3-12. **Detection Rate:** Northern European >99%.

D-bifunctional Protein Deficiency - Gene: HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000414:1-24. **Detection Rate:** Northern European 98%.

Delta-sarcoglycanopathy - **Gene:** SGCD. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000337:2-9. **Detection Rate:** Northern European 96%.

Dihydrolipoamide Dehydrogenase Deficiency - Gene: DLD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000108:1-14. Detection Rate: Northern European >99%.

Dysferlinopathy - **Gene**: DYSF. Autosomal Recessive. Sequencing with copy number analysis. **Exons**: NM_003494:1-55. **Detection Rate**: Northern European 98%.

Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) - Gene: DMD. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_004006:1-79. Detection Rate: Northern European 99%.

ERCC6-related Disorders - Gene: ERCC6. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000124:2-21. **Detection Rate:** Northern European 96%.

ERCC8-related Disorders - Gene: ERCC8. Autosomal Recessive. Sequencing with copy number analysis. **Exons**: NM_000082:1-12. **Detection Rate**: Northern European 97%.

EVC-related Ellis-van Creveld Syndrome - Gene: EVC. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_153717:1-21. **Detection Rate:** Northern European 96%.

EVC2-related Ellis-van Creveld Syndrome - Gene: EVC2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_147127:1-22. **Detection Rate:** Northern European 98%.

Fabry Disease - Gene: GLA. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000169:1-7. Detection Rate: Northern European 98%.

Familial Dysautonomia - Gene: ELP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_003640:2-37. Detection Rate: Northern European >99%.



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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 98%.

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

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

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

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

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

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

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

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%.

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

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

Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - Gene: HBB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000518:1-3. Detection Rate: Northern European >99%. Hereditary Fructose Intolerance - Gene: ALDOB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000035:2-9. Detection Rate: Northern European >99%.

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

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

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

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

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

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

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 Furnpean >99%

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

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

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

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

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

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

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

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

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

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



RESULTS RECIPIENT

SEATTLE SPERM BANK

Attn: Jeffrey Olliffe

NPI: 1306838271

Report Date: 05/27/2021

MALE DONOR 14226

DOB: Ethnicity: Northern European

Barcode:

FEMALE N/A

Primary Hyperoxaluria Type 3 - Gene: HOGA1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_138413:1-7. **Detection Rate:** Northern European >99%.

Pycnodysostosis - **Gene**: CTSK. Autosomal Recessive. Sequencing with copy number analysis. **Exons**: NM_000396:2-8. **Detection Rate**: Northern European >99%.

Pyruvate Carboxylase Deficiency - Gene: PC. Autosomal Recessive. Sequencing with copy number analysis. **Exons**: NM_000920:3-22. **Detection Rate**: Northern European >99%.

Rhizomelic Chondrodysplasia Punctata Type 1 - Gene: PEX7. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000288:1-10. Detection Rate: Northern European >99%.

RTEL1-related Disorders - Gene: RTEL1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_032957:2-35. Detection Rate: Northern European >99%.

Sandhoff Disease - Gene: HEXB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000521:1-14. Detection Rate: Northern European 98%.

Short-chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000017:1-10. Detection Rate: Northern European >99%.

Sjogren-Larsson Syndrome - Gene: ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000382:1-10. **Detection Rate:** Northern European 96%.

SLC26A2-related Disorders - Gene: SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000112:2-3. Detection Rate: Northern European >99%.

Smith-Lemli-Opitz Syndrome - Gene: DHCR7. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001360:3-9. Detection Rate: Northern European >99%.

Spastic Paraplegia Type 15 - Gene: ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_015346:2-42. Detection Rate: Northern European >99%.

Spinal Muscular Atrophy - Gene: SMN1. Autosomal Recessive. Spinal muscular atrophy. Variant (1): SMN1 copy number. Detection Rate: Northern European 95%. Spondylothoracic Dysostosis - Gene: MESP2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001039958:1-2. Detection Rate: Northern European >99%.

TGM1-related Autosomal Recessive Congenital Ichthyosis - Gene: TGM1. Autosomal Recessive. Sequencing with copy number analysis. **Exons**:

NM_000359 2-15. Detection Rate: Northern European >99%.

TPP1-related Neuronal Ceroid Lipofuscinosis - Gene: TPP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000391:1-13. Detection Rate: Northern European >99%.

Tyrosine Hydroxylase Deficiency - Gene: TH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_199292:1-14. **Detection Rate:** Northern European >99%.

Tyrosinemia Type I - Gene: FAH. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000137:1-14. **Detection Rate:** Northern European

Tyrosinemia Type II - **Gene:** TAT. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000353:2-12. **Detection Rate:** Northern European >99%

USH1C-related Disorders - Gene: USH1C. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_005709:1-21. Detection Rate: Northern European >99%.

USH2A-related Disorders - Gene: USH2A. Autosomal Recessive. Sequencing with copy number analysis. **Exons**: NM_206933:2-72. **Detection Rate**: Northern European 98%.

Usher Syndrome Type 3 - Gene: CLRN1. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_174878:1-3. **Detection Rate:** Northern European >99%.

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

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%.



DONOR 14226

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X-linked Juvenile Retinoschisis - Gene: RS1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000330:1-6. Detection Rate: Northern European 98%.

X-linked Myotubular Myopathy - Gene: MTM1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000252:2-15. Detection Rate: Northern European 96%.

X-linked Severe Combined Immunodeficiency - Gene: IL2RG. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000206:1-8. Detection Rate: Northern European >99%.

Xeroderma Pigmentosum Group A - Gene: XPA. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000380:1-6. **Detection Rate:** Northern European >99%.

Xeroderma Pigmentosum Group C - Gene: XPC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_004628:1-16. Detection Rate: Northern European 97%.

Wilson Disease - **Gene:** ATP7B. Autosomal Recessive. Sequencing with copy number analysis. **Exons:** NM_000053:1-21. **Detection Rate:** Northern European >99%.

X-linked Adrenal Hypoplasia Congenita - Gene: NR0B1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000475:1-2. Detection Rate: Northern European 97%.

X-linked Adrenoleukodystrophy - Gene: ABCD1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000033:1-6. Detection Rate: Northern European 77%.

X-linked Alport Syndrome - Gene: COL4A5. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000495:1-51. Detection Rate: Northern European 96%.



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Risk Calculations

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

Disease	DONOR 14226 Residual Risk	Reproductive Risk
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia, HBA1/HBA2-related	Alpha globin status: aa/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	< 1 in 50,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 1,000,000
Argininemia	1 in 12,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 15,000	< 1 in 1,000,000
Aspartylglucosaminuria	< 1 in 50,000	< 1 in 1,000,000
Ataxia with Vitamin E Deficiency	< 1 in 50,000	< 1 in 1,000,000
Ataxia-telangiectasia	1 in 4,200	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 250,000
Autoimmune Polyglandular Syndrome Type 1	1 in 15,000	< 1 in 1,000,000
Autosomal Recessive Osteopetrosis Type 1	1 in 8,900	< 1 in 1,000,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related	1 in 8,100	< 1 in 1,000,000
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	< 1 in 44,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 32,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 42,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	< 1 in 50,000	< 1 in 1,000,000
BCS1L-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Beta-sarcoglycanopathy	1 in 39,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 13,000	1 in 650,000
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 13,000	< 1 in 1,000,000
Canavan Disease	1 in 9,700	< 1 in 1,000,000
Carbamoylphosphate Synthetase I Deficiency	< 1 in 57,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 50,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	1 in 25,000	< 1 in 1,000,000
Cartilage-hair Hypoplasia	< 1 in 50,000	< 1 in 1,000,000
Cerebrotendinous Xanthomatosis	1 in 11,000	< 1 in 1,000,000
Citrullinemia Type 1	1 in 14,000	< 1 in 1,000,000
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 8,600	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
CLN8-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 1 in 1,000,000
COL4A3-related Alport Syndrome	1 in 3,400	< 1 in 1,000,000
COL4A3-related Alport Syndrome COL4A4-related Alport Syndrome	1 in 35,000	< 1 in 1,000,000 < 1 in 1,000,000
Combined Pituitary Hormone Deficiency, PROP1-related	1 in 6,100	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP11B1-related		· · ·
· · · · ·	1 in 8,400 1 in 1,300	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP21A2-related	•	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



MALE

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FEMALE N/A

Disease	DONOR 14226 Residual Risk	Reproductive Risk
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 3,000	1 in 360,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
D-bifunctional Protein Deficiency	1 in 9,000	< 1 in 1,000,000
•	< 1 in 13,000	
Delta-sarcoglycanopathy	•	< 1 in 1,000,000
Dihydrolipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Dysferlinopathy	1 in 11,000	< 1 in 1,000,000
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)	Not calculated	Not calculated
ERCC6-related Disorders	1 in 8,500	< 1 in 1,000,000
ERCC8-related Disorders	< 1 in 16,000	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,800	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	1 in 9,800	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	1 in 220,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 1,000,000
Familial Hyperinsulinism, ABCC8-related	1 in 17,000	< 1 in 1,000,000
Familial Hyperinsulinism, KCNJ11-related	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	1 in 11,000	< 1 in 1,000,000
Fanconi Anemia Complementation Group A	1 in 2,800	< 1 in 1,000,000
Fanconi Anemia, FANCC-related	< 1 in 50,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 16,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Free Sialic Acid Storage Disorders	< 1 in 30,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 37,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,300	< 1 in 1,000,000
Gaucher Disease	1 in 260	
		1 in 110,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 2,500	1 in 260,000
GLB1-related Disorders	1 in 17,000	< 1 in 1,000,000
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
Glycine Encephalopathy, AMT-related	1 in 26,000	< 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 20,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 20,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Si Disease)	ckle Cell 1 in 3,700	1 in 560,000
Hereditary Fructose Intolerance	1 in 7,900	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 30,000	< 1 in 1,000,000
HMG-CoA Lyase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	< 1 in 1,000,000
Homocystinuria, CBS-related	1 in 9,400	< 1 in 1,000,000
Hydrolethalus Syndrome	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia	1 in 30,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 32,000	
		< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMB3-related	1 in 32,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 14,000	< 1 in 1,000,000
Leigh Syndrome, French-Canadian Type	< 1 in 50,000	< 1 in 1,000,000
Lipoid Congenital Adrenal Hyperplasia	< 1 in 50,000	< 1 in 1,000,000
Lysosomal Acid Lipase Deficiency	1 in 14,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 39,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ib	1 in 39,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 16,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 4,400	1 in 790,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	< 1 in 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 16,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 1,000,000
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MALE DONOR 14226

DOB: Ethnicity: Northern European

Barcode:

FEMALE N/A

Pi	DONOR 14226	Donas donation Dist
Disease	Residual Risk	Reproductive Risk
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
Mucolipidosis III Gamma	< 1 in 20,000	< 1 in 1,000,000
Mucolipidosis IV	< 1 in 50,000	< 1 in 1,000,000
Mucopolysaccharidosis Type I	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type II	< 1 in 1,000,000	1 in 300,000
Mucopolysaccharidosis Type IIIA	1 in 19,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 27,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	< 1 in 50,000	< 1 in 1,000,000
Muscular Dystrophy, LAMA2-related	1 in 5,700	< 1 in 1,000,000
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	1 in 35,000	< 1 in 1,000,000
Neuronal Ceroid Lipofuscinosis, CLN6-related	1 in 20,000	< 1 in 1,000,000
Niemann-Pick Disease Type C1	1 in 19,000	< 1 in 1,000,000
Niemann-Pick Disease Type C2	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-related	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	1 in 16,000	< 1 in 1,000,000
Ornithine Transcarbamylase Deficiency	< 1 in 1,000,000	1 in 140,000
PCCA-related Propionic Acidemia	1 in 4,200	< 1 in 1,000,000
PCCB-related Propionic Acidemia	1 in 22,000	< 1 in 1,000,000
PCDH15-related Disorders	1 in 3,300	< 1 in 1,000,000
Pendred Syndrome	1 in 8,200	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 1	1 in 16,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 3	1 in 44,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 4	1 in 9,300	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 5	< 1 in 71,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 6	< 1 in 50,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 4,800	1 in 940,000
POMGNT-related Disorders	< 1 in 12,000	< 1 in 1,000,000
Pompe Disease	1 in 4,000	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	1 in 7,700	< 1 in 1,000,000
Primary Carnitine Deficiency	1 in 11,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 17,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	1 in 13,000	< 1 in 1,000,000
Pycnodysostosis	1 in 43,000	< 1 in 1,000,000
Pyruvate Carboxylase Deficiency	1 in 25,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	< 1 in 1,000,000
RTEL1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Sandhoff Disease	NM_000521.3(HEXB) c.1250C>T(P417L) heter	
Short-chain Acyl-CoA Dehydrogenase Deficiency	1 in 11,000	< 1 in 1,000,000
Sjogren-Larsson Syndrome	< 1 in 12,000	< 1 in 1,000,000
SLC26A2-related Disorders	1 in 16,000	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 9,400	< 1 in 1,000,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770	1 in 110,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	< 1 in 1,000,000
Tyrosine Hydroxylase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 16,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 30,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 4,100	< 1 in 1,000,000
Usher Syndrome Type 3	1 in 41,000	< 1 in 1,000,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency	1 in 18,000	< 1 in 1,000,000
very-long-chain acyr-coa benyurogenase bendency	1 111 10,000	< 1 III 1,000,000



MALE DONOR 14226

DOB: Ethnicity: Northern European

Barcode:

FEMALE N/A

Disease	DONOR 14226 Residual Risk	Reproductive Risk
Wilson Disease	1 in 6,500	< 1 in 1,000,000
X-linked Adrenal Hypoplasia Congenita	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 90,000	1 in 42,000
X-linked Alport Syndrome	Not calculated	Not calculated
X-linked Juvenile Retinoschisis	< 1 in 1,000,000	1 in 40,000
X-linked Myotubular Myopathy	Not calculated	Not calculated
X-linked Severe Combined Immunodeficiency	< 1 in 1,000,000	1 in 200,000
Xeroderma Pigmentosum Group A	< 1 in 50,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group C	1 in 7,300	< 1 in 1,000,000

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Patient Information:

14226, Donor DOB: Sex: M

MR#: Patient#: FT-PT9242079 Accession: FT-7702741

Test#: FT-TS15358818 Order#: FT-OR15330282 Ext Test#: WP-TTS19419525 Ext Order#: WP-TOR19404526 Specimen Type: Saliva

Collected: Sep 30,2025 Received Date: Oct 03,2025 Authorized Date: Oct 07,2025 Physician:

Kuan, James

ATTN: SSB Genetics, Dept San Diego Sperm Bank 4915 25th Avenue NE, Ste 204W Seattle, WA 98105

Phone: (206) 588-1484

Fax:

Laboratory:

Fulgent Therapeutics LLC

CAP#: 8042697 CLIA#: 05D2043189 Laboratory Director: Dr. Amar Jariwala

Report Date: Oct 25,2025

Final Report

TEST PERFORMED

Known Mutation / Site-Specific Testing - Targeted testing for 1 variant was performed in the submitted specimen.

RESULTS

1 out of 1 variant(s) were detected.

VARIANTS TESTED:

	GENE INFO	VARIANT INFO			
Gene	Inheritance	Variant	Zygosity	Classification	Status
CLCNKB	Autosomal Recessive	NM_000085.5:c.968+1G>A (p.?)	Heterozygous	Pathogenic	Detected

NOTES AND RECOMMENDATIONS:

- Gene specific notes and limitations may be present. See below.
- These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; https://www.nsgc.org)

Patient: 14226, Donor; Sex: M; DOB: MR#:

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INTERPRETATION:

ABOUT CLCNKB

Biallelic pathogenic variants in CLCNKB are associated with Bartter syndrome type 3, characterized by impaired salt reabsorption in the thick ascending loop of Henle with pronounced salt wasting, hypokalemic metabolic alkalosis, and hypercalciuria (PubMed: 9326936, 22282380, 16785747, 17061123; OMIM: 607364). Digenic inheritance of Bartter syndrome type 4B has been reported involving biallelic pathogenic variants or deletions in the CLCNKB gene in addition to biallelic mutations or deletions in the CLCNKA gene (PubMed: 15044642, 18310267; OMIM: 613090).

CLCNKB NM_000085.5:c.968+1G>A (p.?)

DETECTED - Classification: Pathogenic

Targeted testing of NM_000085.5:c.968+1G>A (p.?), in the CLCNKB gene, was performed in this specimen. The test results reported here indicate that the variant was detected as heterozygous in this specimen. This variant is in the dbSNP database: rs201204502. This variant is predicted to disrupt the canonical splice donor site for exon 10. Functional analysis demonstrating aberrant splicing or aberrant regulation would be required to further refine the categorization of this variant. This intronic variant alters the highly conserved consensus sequence for the canonical splice site and is predicted by multiple splice site prediction tools queried to abolish canonical splice activity. This variant is expected to result in altered function of the gene product as a result of aberrant splicing. There's sufficient evidence that loss of function in this gene is a known disease mechanism for Bartter syndrome (PubMed: 29254190, 32857947, 31834604, 28381550). This splicing variant has previously been reported in association with Bartter syndrome type III and congenital anomalies of the kidney and urinary tract, and has been shown to segregate with disease in one family (PubMed: 23164417). Additionally, this variant has been identified in the compound heterozygous state in an individual with Bartter syndrome type 3 by another laboratory. This variant is classified as a "Disease Mutation" (DM) in the Human Gene Mutation Database (HGMD). This variant has been observed at a frequency of less than 0.01% (16/250750 alleles) in the Broad gnomAD dataset. The highest allele frequency that this variant has been observed at in any sub-population with available data is 0.01% in the European (Non-Finnish) population. There are no homozygous control individuals for this variant. The Broad Institute gnomAD database (>120,000 Individuals with no known severe, pediatric onset disease) was used for this analysis.

Patient: 14226, Donor; Sex: M; DOB: MR#:

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GENES TESTED:

Known Mutation / Site-Specific Testing

1 genes tested CLCNKB

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). Methodology - Next Generation Sequencing (NGS), Sanger Sequencing, quantitative PCR (qPCR), repeat-primed PCR (rpPCR) or multiplex ligation-dependent probe amplification (MLPA) is selected by the laboratory to provide optimal results. (Panel: FT-TP00142v1)

If NGS was performed: DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37 / hg19), variants were detected in regions of at least 10x coverage. The known mutation genomic loci requested are evaluated for the presence or absence of variation compared to the human genome reference sequence. Bioinformatics: FPLMv2.0 was used to generate variant calls for this test.

If Sanger Sequencing was performed: DNA was amplified for the target region and sequenced bi-directionally using an ABI 3730XL instrument. The data was analyzed against the reference gene sequence and the known variant position as requested.

If qPCR was performed: DNA was amplified for the target region and quantified using a QuantStudio 6 instrument. The data is compared to control genes and control individuals for the targets as requested.

If rpPCR was performed: This analysis is performed by repeat-primed PCR (rpPCR) and amplicon length analysis. The scope of this assay is limited to repeat expansion analysis of the specified gene. Gene sequencing and deletion/duplication analysis are not included in this assay. This analysis does not include methylation studies.

If MLPA was performed: DNA was amplified for the target regions and quantified using probesets using kits from MRC-Holland and an ABI 3730 instrument. The data is compared to control genes and control individuals for the targets as requested.

LIMITATIONS:

All laboratory tests have limitations. These results assume that the specimen received in the laboratory belongs to the named individual and that no mix-up or co-mingling of specimens has occurred. Positive results do not imply that there are no other pathogenic alterations in the patient's genome, and negative results do not rule out a genetic cause for the indication for testing. This assay assumes that any stated familial relationships are accurate. This assay is not designed or validated for the detection of somatic mosaicism or somatic mutations. This assay will only analyze the variant(s) requested. It is possible that the nomenclature for the variants tested may be different from the requested variants due to nomenclature differences in different isoforms of the gene. It is very important to provide us the isoform (NM number) of the gene for every variant to be tested. Result interpretation assumes that the human reference sequences are correct at the queried loci. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (https://www.genenames.org) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the collected information available at the time of reporting; additional information may exist in the future which will not be represented. Rarely, due to systematic chemical or computational issues, or human error, DNA variants may be missed. If a positive familial control specimen is not provided or available, rare errors may occur.

Gene Specific Notes and Limitations

No gene specific limitations apply to the genes on the tested panel.

SIGNATURE:

Zhenbin Chen, Ph.D., CGMB, FACMG on 10/25/2025

Laboratory Director, Fulgent

DISCLAIMER:

This test was developed, performed, and its performance characteristics determined by **Fulgent Therapeutics LLC** (CAP# 8042697, CLIA# 05D2043189), 4399 Santa Anita Ave., El Monte, CA 91731. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at (626) 350-0537 or info@fulgentgenetics.com. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.

Patient: 14226, Donor; Sex: M; DOB: MR#:

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