



RESULTS RECIPIENT
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Report Date: 09/30/2019

MALE
DONOR 10407
DOB: [REDACTED]
Ethnicity: Northern European
Sample Type: EDTA Blood
Date of Collection: 09/20/2019
Date Received: 09/21/2019
Date Tested: 09/30/2019
Barcode: 11004212735019
Accession ID:
CSLW4EXQDFWUEGA
Indication: Egg or sperm donor

FEMALE
N/A

Foresight® Carrier Screen

POSITIVE: CARRIER

ABOUT THIS TEST

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

RESULTS SUMMARY

Risk Details	DONOR 10407	Partner
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel (175 conditions tested)	N/A
POSITIVE: CARRIER Hereditary Fructose Intolerance Reproductive Risk: 1 in 320 Inheritance: Autosomal Recessive	+ CARRIER* NM_000035.3(ALDOB):c. 448G>C(A150P) heterozygote	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".

*Carriers generally do not experience symptoms.

No disease-causing mutations were detected in any other gene tested. A complete list of all conditions tested can be found on page 6.

CLINICAL NOTES

- None

NEXT STEPS

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

POSITIVE: CARRIER

Hereditary Fructose Intolerance

Reproductive risk: 1 in 320

Risk before testing: 1 in 25,000

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

Patient	DONOR 10407	No partner tested
Result	⊕ Carrier	N/A
Variant(s)	NM_000035.3(ALDOB):c.448G>C(A150P) heterozygote	N/A
Methodology	Sequencing with copy number analysis	N/A
Interpretation	This individual is a carrier of hereditary fructose intolerance. Carriers generally do not experience symptoms.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000035:2-9.	N/A

What Is Hereditary Fructose Intolerance (HFI)?

Hereditary fructose intolerance (HFI) is a disorder caused by mutations in the *ALDOB* gene that is characterized by an inability to break down fructose, a common sugar found in fruit and many other foods. When an individual with HFI consumes fructose, the result is low blood sugar (hypoglycemia) and a buildup of toxic substances in the liver.

The first symptoms of HFI usually appear when a child is first introduced to formula or foods containing fructose or the related sugars sucrose and sorbitol (a sugar substitute). Symptoms that may appear include irritability, upset stomach, vomiting, sweating, and/or sleepiness. If unrecognized and untreated, children with HFI will fail to grow at a normal rate, may develop a yellowing of the skin and whites of the eyes (jaundice), and have enlargement of the liver and spleen (hepatosplenomegaly). Without treatment, HFI can eventually lead to serious liver disease, hypoglycemic shock, seizures, and kidney or liver failure. In extreme cases, it can be fatal. For this reason, early detection and treatment is critical.

Symptoms of HFI can vary from mild to severe. Individuals with HFI often show an aversion to sweets and fruit, and those with mild HFI may, therefore, be protected from some of the symptoms they would otherwise experience. In addition, those with a severe course of the disease may develop serious liver disease later in life, even with a careful diet.

How Common Is HFI?

The estimated prevalence of HFI is approximately 1 in 20,000 to 1 in 30,000 individuals worldwide. However, the condition may be more or less common among certain ethnic groups.

How Is HFI Treated?

Treatment for HFI involves strict control of diet, eliminating all foods or products (for example, medicines or vitamins) containing fructose, sucrose, or sorbitol. With a carefully managed diet, individuals with HFI can be symptom-free, although symptoms will quickly return upon consuming fructose, sucrose, or sorbitol. In cases where liver disease has progressed to a life-threatening stage, liver transplantation is possible.

What Is the Prognosis for an Individual with HFI?

Without elimination of all fructose, sucrose, and sorbitol, HFI can be life threatening. Continued consumption of these sugars can lead to hypoglycemic shock, seizures, coma, serious liver disease, liver or kidney failure, and potentially death. With a careful diet, however, individuals with HFI may be symptom-free, have normal growth and development, and have a normal life expectancy. The earlier the condition is diagnosed and the diet adjusted, the less damage is done to the liver and kidneys and the better the overall prognosis. Early detection and diet modification are also important for age-appropriate growth. In a minority of individuals who have a severe form of HFI, liver disease may still develop, despite a careful diet.

Methods and Limitations

DONOR 10407 [Foresight Carrier Screen]: Sequencing with copy number analysis, spinal muscular atrophy, and analysis of homologous regions.

Sequencing with copy number analysis

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

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect. Detection rates only account for analytical sensitivity and certain variants that have been previously described in the literature may not be reported if there is insufficient evidence for pathogenicity. Detection rates do not account for the disease-specific rates of de novo mutations.

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

Spinal muscular atrophy

Targeted copy number analysis is used to determine the copy number of exon 7 of the *SMN1* gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of *SMN1* are carriers with two *SMN1* genes on one chromosome and a *SMN1* deletion on the other chromosome. This is more likely in individuals who have 2 copies of the *SMN1* gene and are positive for the g.27134T>G SNP, which affects the reported residual risk; Ashkenazi Jewish or Asian patients with this genotype have a high post-test likelihood of being carriers for SMA and are reported as carriers. The g.27134T>G SNP is only reported in individuals who have 2 copies of *SMN1*.

Analysis of homologous regions

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

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

Limitations

In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. This test is designed to detect and report germline alterations. While somatic variants present at low levels may be detected, these may not be reported. 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 Myriad Women's Health, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's evaluation. CLIA Number: **#05D1102604**.

Resources

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

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

SENIOR LABORATORY DIRECTOR



Jack Ji, PhD, FACMG

Report content approved by Lulu Mao, PhD, DABMGG on Sep 30, 2019

Conditions Tested

11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia - Gene: CYP11B1.

Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000497:1-9. **Detection Rate:** Northern European 94%.

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

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

Adenosine Deaminase Deficiency - Gene: ADA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000022:1-12. **Detection Rate:** Northern European >99%.

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

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

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

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

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

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

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

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

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

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

Ataxia-telangiectasia - Gene: ATM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000051:2-63. **Detection Rate:** Northern European 98%.

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

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

Autosomal Recessive Osteopetrosis Type 1 - Gene: TCIRG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006019:2-20. **Detection Rate:** Northern European >99%.

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

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay - Gene: SACS.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

COL4A3-related Alport Syndrome - Gene: COL4A3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000091:1-52. **Detection Rate:** Northern European 97%.

COL4A4-related Alport Syndrome - Gene: COL4A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000092:2-48. **Detection Rate:** Northern European 98%.

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

Congenital Adrenal Hyperplasia, CYP21A2-related - Gene: CYP21A2. Autosomal Recessive. Analysis of homologous regions. Variants (13): CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs*21, I173N, L308Ffs*6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V281L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Northern European 96%.

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

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

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

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

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

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

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

Delta-sarcoglycanopathy - Gene: SGCD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000337:2-9. **Detection Rate:** Northern European 99%.

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

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

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

ERCC6-related Disorders - Gene: ERCC6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000124:2-21. **Detection Rate:** Northern European 99%.

ERCC8-related Disorders - Gene: ERCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000082:1-12. **Detection Rate:** Northern European 95%.

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

EVC2-related Ellis-van Creveld Syndrome - Gene: EVC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_147127:1-22. **Detection Rate:** Northern European >99%.

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

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

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

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

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

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

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

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

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

Gamma-sarcoglycanopathy - Gene: SGCG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000231:2-8. **Detection Rate:** Northern European 88%.

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

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

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

GLDC-related Glycine Encephalopathy - Gene: GLDC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000170:1-25. **Detection Rate:** Northern European 94%.

Glutaric Acidemia, GCDH-related - Gene: GCDH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000159:2-12. **Detection Rate:** Northern European >99%.

Glycogen Storage Disease Type Ia - Gene: G6PC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000151:1-5. **Detection Rate:** Northern European >99%.

Glycogen Storage Disease Type Ib - Gene: SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001164277 3-11. **Detection Rate:** Northern European >99%.

Glycogen Storage Disease Type III - Gene: AGL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000642:2-34. **Detection Rate:** Northern European >99%.

GNE Myopathy - Gene: GNE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001128227:1-12. **Detection Rate:** Northern European >99%.

GNPTAB-related Disorders - Gene: GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_024312:1-21. **Detection Rate:** Northern European >99%.

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

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

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

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

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

HMG-CoA Lyase Deficiency - Gene: HMGCL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000191:1-9. **Detection Rate:** Northern European 98%.

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

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

Hydroletharus Syndrome - Gene: HYL1. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_145014:4. **Detection Rate:** Northern European >99%.

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

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

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

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

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

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

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

LAMA2-related Muscular Dystrophy - Gene: LAMA2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000426:1-65. **Detection Rate:** Northern European >99%.

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

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

Lysosomal Acid Lipase Deficiency - Gene: LIPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000235:2-10. **Detection Rate:** Northern European >99%.

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

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

Maple Syrup Urine Disease Type II - Gene: DBT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001918:1-11. **Detection Rate:** Northern European 96%.

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

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

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

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

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

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

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

Mucopolidosis III Gamma - Gene: GNPTG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_032520:1-11. **Detection Rate:** Northern European >99%.

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

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

Mucopolysaccharidosis Type II - Gene: IDS. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000202:1-9. **Detection Rate:** Northern European 88%.

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

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

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

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

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

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

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

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

Niemann-Pick Disease Type C1 - Gene: NPC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000271:1-25. **Detection Rate:** Northern European >99%.

Niemann-Pick Disease Type C2 - Gene: NPC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_006432:1-5. **Detection Rate:** Northern European >99%.

Niemann-Pick Disease, SMPD1-related - Gene: SMPD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000543:1-6. **Detection Rate:** Northern European >99%.

Nijmegen Breakage Syndrome - Gene: NBN. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_002485:1-16. **Detection Rate:** Northern European >99%.

Ornithine Transcarbamylase Deficiency - Gene: OTC. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000531:1-10. **Detection Rate:** Northern European 97%.

PCCA-related Propionic Acidemia - Gene: PCCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000282:1-24. **Detection Rate:** Northern European 95%.

PCCB-related Propionic Acidemia - Gene: PCCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000532:1-15. **Detection Rate:** Northern European >99%.

PCDH15-related Disorders - Gene: PCDH15. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_033056:2-33. **Detection Rate:** Northern European 93%.

Pendred Syndrome - Gene: SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000441:2-21. **Detection Rate:** Northern European >99%.

Peroxisome Biogenesis Disorder Type 1 - Gene: PEX1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000466:1-24. **Detection Rate:** Northern European >99%.

Peroxisome Biogenesis Disorder Type 3 - Gene: PEX12. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000286:1-3. **Detection Rate:** Northern European >99%.

Peroxisome Biogenesis Disorder Type 4 - Gene: PEX6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000287:1-17. **Detection Rate:** Northern European 97%.

Peroxisome Biogenesis Disorder Type 5 - Gene: PEX2. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM_000318:4. **Detection Rate:** Northern European >99%.

Peroxisome Biogenesis Disorder Type 6 - Gene: PEX10. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_153818:1-6. **Detection Rate:** Northern European >99%.

Phenylalanine Hydroxylase Deficiency - Gene: PAH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000277:1-13. **Detection Rate:** Northern European >99%.

POMGNT-related Disorders - Gene: POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_017739:2-22. **Detection Rate:** Northern European 96%.

Pompe Disease - Gene: GAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000152:2-20. **Detection Rate:** Northern European 98%.

PPT1-related Neuronal Ceroid Lipofuscinosis - Gene: PPT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000310:1-9. **Detection Rate:** Northern European >99%.

Primary Carnitine Deficiency - Gene: SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_003060:1-10. **Detection Rate:** Northern European >99%.

Primary Hyperoxaluria Type 1 - Gene: AGXT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000030:1-11. **Detection Rate:** Northern European >99%.

Primary Hyperoxaluria Type 2 - Gene: GRHPR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_012203:1-9. **Detection Rate:** Northern European >99%.

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

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

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

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

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

Salla Disease - Gene: SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_012434:1-11. **Detection Rate:** Northern European 98%.

Sandhoff Disease - Gene: HEXB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000521:1-14. **Detection Rate:** Northern European >99%.

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

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

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

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

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

Spinal Muscular Atrophy - Gene: SMN1. Autosomal Recessive. Spinal muscular atrophy. Variant (1): SMN1 copy number. **Detection Rate:** Northern European 95%.

Spondyl thoracic Dysostosis - Gene: MESP2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_001039958:1-2. **Detection Rate:** Northern European >99%.

TGM1-related Autosomal Recessive Congenital Ichthyosis - Gene: TGM1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000359 2-15. **Detection Rate:** Northern European >99%.

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

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

Tyrosinemia Type I - Gene: FAH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000137:1-14. **Detection Rate:** Northern European >99%.

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

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

USH2A-related Disorders - Gene: USH2A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_206933:2-72. **Detection Rate:** Northern European 94%.

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

Very-long-chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADVL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM_000018:1-20. **Detection Rate:** Northern European >99%.



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Jeffrey Olliffe
NPI: 1306838271
Report Date: 09/30/2019

MALE	FEMALE
DONOR 10407	N/A
DOB: [REDACTED]	
Ethnicity: Northern European	
Barcode: 11004212735019	

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

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

X-linked Alport Syndrome - Gene: COL4A5. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000495:1-51. **Detection Rate:** Northern European 95%.

X-linked Congenital Adrenal Hypoplasia - Gene: NR0B1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000475:1-2. **Detection Rate:** Northern European 99%.

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

X-linked Myotubular Myopathy - Gene: MTM1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM_000252:2-15. **Detection Rate:** Northern European 98%.

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

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

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

Risk Calculations

Below are the risk calculations for all conditions tested. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's future children could inherit each disease. These risks are inherent to all carrier screening tests, may vary by ethnicity, are predicated on a negative family history and are present even after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

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

Disease	DONOR 10407 Residual Risk	Reproductive Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 3,800	< 1 in 1,000,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 1,000,000
ABCC8-related Familial Hyperinsulinism	1 in 17,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia	Alpha globin status: aa/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	1 in 45,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 1,000,000
AMT-related Glycine Encephalopathy	1 in 22,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 1,000,000
Argininemia	< 1 in 17,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 13,000	< 1 in 1,000,000
Aspartylglucosaminuria	< 1 in 50,000	< 1 in 1,000,000
Ataxia with Vitamin E Deficiency	< 1 in 50,000	< 1 in 1,000,000
Ataxia-telangiectasia	1 in 11,000	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 600,000
Autoimmune Polyglandular Syndrome Type 1	1 in 15,000	< 1 in 1,000,000
Autosomal Recessive Osteopetrosis Type 1	1 in 35,000	< 1 in 1,000,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related	1 in 8,100	< 1 in 1,000,000
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	< 1 in 44,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 32,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 42,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	< 1 in 50,000	< 1 in 1,000,000
BCS1L-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Beta-sarcoglycanopathy	1 in 39,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 13,000	1 in 650,000
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 13,000	< 1 in 1,000,000
Canavan Disease	1 in 9,700	< 1 in 1,000,000
Carbamoylphosphate Synthetase I Deficiency	< 1 in 57,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 50,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	1 in 25,000	< 1 in 1,000,000
Cartilage-hair Hypoplasia	< 1 in 50,000	< 1 in 1,000,000
Cerebrotendinous Xanthomatosis	1 in 11,000	< 1 in 1,000,000
Citrullinemia Type 1	1 in 14,000	< 1 in 1,000,000
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 8,600	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
CLN6-related Neuronal Ceroid Lipofuscinosis	1 in 43,000	< 1 in 1,000,000
CLN8-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 1 in 1,000,000
COL4A3-related Alport Syndrome	1 in 6,200	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 12,000	< 1 in 1,000,000
Combined Pituitary Hormone Deficiency, PROP1-related	1 in 6,100	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP21A2-related	1 in 1,300	1 in 280,000
Congenital Disorder of Glycosylation Type Ia	1 in 16,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation, MPI-related	< 1 in 50,000	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 1,000,000

Disease	DONOR 10407 Residual Risk	Reproductive Risk
Cystic Fibrosis	1 in 3,000	1 in 360,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
D-bifunctional Protein Deficiency	1 in 9,000	< 1 in 1,000,000
Delta-sarcoglycanopathy	< 1 in 40,000	< 1 in 1,000,000
Dihydrolipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Dysferlinopathy	1 in 11,000	< 1 in 1,000,000
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)	Not calculated	Not calculated
ERCC6-related Disorders	1 in 26,000	< 1 in 1,000,000
ERCC8-related Disorders	< 1 in 9,900	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,500	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	< 1 in 50,000	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	1 in 80,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	< 1 in 50,000	< 1 in 1,000,000
Fanconi Anemia Complementation Group A	1 in 2,800	< 1 in 1,000,000
Fanconi Anemia, FANCC-related	< 1 in 50,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 16,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 10,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,000	< 1 in 1,000,000
Gaucher Disease	1 in 260	1 in 110,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 2,500	1 in 260,000
GLB1-related Disorders	1 in 19,000	< 1 in 1,000,000
GLDC-related Glycine Encephalopathy	1 in 2,800	< 1 in 1,000,000
Glutaric Acidemia, GCDH-related	1 in 16,000	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 18,000	< 1 in 1,000,000
Glycogen Storage Disease Type Ib	1 in 35,000	< 1 in 1,000,000
Glycogen Storage Disease Type III	1 in 16,000	< 1 in 1,000,000
GNE Myopathy	1 in 23,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 32,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 20,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 3,100	1 in 390,000
Hereditary Fructose Intolerance	A150P heterozygote [†]	1 in 320
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related	< 1 in 50,000	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 30,000	< 1 in 1,000,000
HMG-CoA Lyase Deficiency	< 1 in 33,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	< 1 in 1,000,000
Homocystinuria, CBS-related	1 in 9,400	< 1 in 1,000,000
Hydrolethalus Syndrome	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia	1 in 27,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 32,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 50,000	< 1 in 1,000,000
KCNJ11-related Familial Hyperinsulinism	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 14,000	< 1 in 1,000,000
LAMA2-related Muscular Dystrophy	1 in 34,000	< 1 in 1,000,000
Leigh Syndrome, French-Canadian Type	< 1 in 50,000	< 1 in 1,000,000
Lipoid Congenital Adrenal Hyperplasia	< 1 in 50,000	< 1 in 1,000,000
Lysosomal Acid Lipase Deficiency	1 in 18,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 42,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ib	1 in 39,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 13,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 4,400	1 in 790,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	< 1 in 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 16,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblB Type	1 in 48,000	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 16,000	< 1 in 1,000,000
MKS1-related Disorders	< 1 in 50,000	< 1 in 1,000,000

Disease	DONOR 10407 Residual Risk	Reproductive Risk
Mucopolipidosis III Gamma	< 1 in 50,000	< 1 in 1,000,000
Mucopolipidosis IV	< 1 in 50,000	< 1 in 1,000,000
Mucopolysaccharidosis Type I	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type II	1 in 600,000	1 in 150,000
Mucopolysaccharidosis Type IIIA	1 in 12,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 25,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	1 in 37,000	< 1 in 1,000,000
MUT-related Methylmalonic Acidemia	1 in 26,000	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	1 in 1,200	1 in 400,000
Nephrotic Syndrome, NPHS1-related	< 1 in 50,000	< 1 in 1,000,000
Nephrotic Syndrome, NPHS2-related	1 in 35,000	< 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
Salla Disease	< 1 in 30,000	< 1 in 1,000,000
Sandhoff Disease	1 in 32,000	< 1 in 1,000,000
Short-chain Acyl-CoA Dehydrogenase Deficiency	1 in 11,000	< 1 in 1,000,000
Sjogren-Larsson Syndrome	< 1 in 12,000	< 1 in 1,000,000
SLC26A2-related Disorders	1 in 16,000	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 9,400	< 1 in 1,000,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770	1 in 110,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	< 1 in 1,000,000
Tyrosine Hydroxylase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 16,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 35,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 2,200	< 1 in 1,000,000
Usher Syndrome Type 3	1 in 41,000	< 1 in 1,000,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency	1 in 18,000	< 1 in 1,000,000
Wilson Disease	1 in 8,600	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 90,000	1 in 42,000
X-linked Alport Syndrome	Not calculated	Not calculated
X-linked Congenital Adrenal Hypoplasia	< 1 in 1,000,000	< 1 in 1,000,000



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Jeffrey Olliffe
NPI: 1306838271
Report Date: 09/30/2019

MALE
DONOR 10407
DOB: [REDACTED]
Ethnicity: Northern European
Barcode: 11004212735019

FEMALE
N/A

Disease	DONOR 10407 Residual Risk	Reproductive Risk
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

DOB
[REDACTED]

Accession
3076690

Submitter Patient ID(s) 11717187

Sample

Source: OraCollect Buccal
Date Collected: 8/8/2024
Date Received: 8/9/2024

Testing

Date Started: 8/19/2024
Date Reported: 9/5/2024

Provider

Account #: ZZ280
Kuan, James

Test(s) Requested

Targeted Variant Testing

Clinical Indications

Targeted testing for the variant(s) below was requested.

The requested variant(s) may have been previously reported using alternate nomenclature as gene names and variant nomenclature may differ among laboratories or change over time.

Result: Heterozygous Carrier

Gene	Mode of Inheritance	Variant	Reference Sequence	Classification	Result
NAGA	Autosomal recessive	c.973 G>A p.(E325K)	NM_000262.2	Pathogenic Variant	Heterozygous

Interpretation

This donor is a heterozygous carrier for a variant that was previously identified in the proband. If the other biological parent is a heterozygous carrier for the p.(E325K) variant, then this confirms that the variant(s) are present on opposite alleles (in trans) in the proband.

Recommendation

- Genetic counseling is recommended to discuss the implications of these results for this individual and family members.
- Targeted carrier testing may be considered for family members for the variant identified in this individual.

NAGA

GENE SUMMARY

The NAGA gene encodes alpha-N-acetylgalactosaminidase, a lysosomal enzyme that cleaves alpha-N-acetylgalactosamine residues from glycopeptides and glycolipids (PMID: 19683538). Variants in the NAGA gene are associated with autosomal recessive alpha-N-acetylgalactosaminidase (NAGA) deficiency. This is a very rare lysosomal storage disorder with three phenotypes. Type I, also known as Schindler disease type I, is a severe infantile-onset neurodegenerative disorder (PMID: 19683538). Type II, also known as Kanzaki disease, is an adult-onset condition characterized by angiokeratoma corporis diffusum and mild intellectual disability (MIM 609241). Type III, also known as Schindler disease type III, is an intermediate disorder characterized by a spectrum of symptoms including seizures, autistic disorders, and cardiomyopathy (PMID: 19683538). To date, most pathogenic variants reported in the NAGA gene have been missense variants, however truncating variants have also been reported (HGMD).

p.(Glu325Lys) (GAA>AAA): c.973 G>A in exon 8 of the NAGA gene (NM_000262.2)

- Reported previously in both the homozygous and compound heterozygous states in individuals with alpha-N-acetylgalactosaminidase

DOB

[REDACTED]

Accession

3076690

Submitter Patient ID(s) 11717187

-
- deficiency referred for genetic testing at GeneDx and in published literature (PMID: 2243144, 11313741, 8782044)
- Functional studies indicate this variant results in loss of alpha-N-acetylgalactosaminidase activity (PMID: 2243144)
 - Observed in large population cohorts (gnomAD; internal data) and no individuals are reported as homozygous

We interpret this as a Pathogenic Variant.

Additional Comments

Variants elsewhere in the evaluated gene would not be identified by this targeted analysis. Variant classification may change over time. If the current variant classification differs from that of other family members tested at GeneDx, an updated report will be sent to their providers.

If a positive control was not provided or the variant was not previously identified in this family at GeneDx, there may be rare circumstances in which the variant is present but is not detected.

Methods

Using genomic DNA from the submitted specimen, the relevant portion of the requested gene was PCR amplified and capillary sequencing was performed. Bi-directional sequence was assembled, aligned to reference gene sequences based on human genome build GRCh37/UCSC hg19 and analyzed for known familial sequence variant(s). Sequence alterations were reported according to the Human Genome Variation Society (HGVS) nomenclature guidelines. The methods used by GeneDx are expected to be greater than 99% sensitive in detecting variants identifiable by sequencing. Available evidence for variant classification may change over time and reported variant(s) may be reclassified according to the ACMG/AMP Standards and Guidelines (PMID: 25741868), which may lead to issuing a revised report.

Disclaimer

Genetic testing using the methods applied at GeneDx is expected to be highly accurate. Normal findings do not rule out the diagnosis of a genetic disorder since some genetic abnormalities may be undetectable by this test. Unless otherwise indicated, the methods used cannot reliably detect deletions of 20bp to 500bp in size, or insertions of 10bp to 500bp in size. Sequencing cannot detect low-level mosaicism. Copy number assessment methods cannot reliably detect mosaicism and cannot identify balanced chromosome aberrations. Regions of certain genes have inherent sequence properties (for example: repeat, homology, or pseudogene regions, high GC content, rare polymorphisms) that yield suboptimal data, potentially impairing accuracy of the results. Inaccurate results may occur in the setting of allogeneic bone marrow/stem cell transplantation, active or chronic hematologic conditions, recent blood transfusion, suboptimal DNA quality, or in other rare circumstances. DNA extracted at other laboratories may negatively affect test performance. Rarely, incidental findings of large chromosomal rearrangements outside the gene of interest may be identified. As the ability to detect genetic variants and naming conventions can differ among laboratories, rare false negative results may occur when no positive control is provided for testing of a specific variant identified at another laboratory. In addition, the chance of an erroneous result due to laboratory errors incurred during any phase of testing cannot be completely excluded. The clinical sensitivity of this test depends in part on the patient's clinical presentation and is expected to be highest for individuals with a clearly defined phenotype and/or family history. Interpretations are made with the assumption that any clinical information provided, including family relationships, are accurate. Consultation with a genetics professional is recommended for interpretation of results. This test was developed and its performance characteristics determined by GeneDx.

This test has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. The test is used for clinical purposes and should not be regarded as investigational or for research. The laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing.

References

Lek et al. (2016) Nature 536 (7616):285-91 (PMID: 27535533); Stenson et al. (2014) Human genetics 133 (1):1-9 (PMID: 24077912); Landrum et al. (2016) Nucleic Acids Res. 44 (D1):D862-8 (PMID: 26582918); Lott et al. (2013) Curr Protoc Bioinformatics 44 :1.23.1-26 (PMID: 25489354); Richards et al. (2015) Genetics In Medicine: 17 (5):405-24 (PMID: 25741868);

Report Electronically Signed By

Adam J Guenzel PhD, FACMG

Assistant Director

DOB
[REDACTED]

Accession
3076690

Submitter Patient ID(s) 11717187

Variant Table

Gene: Coding DNA	NAGA: c.973 G>A
Variant (Protein)	p.(Glu325Lys) ((E325K))
Classification	Pathogenic Variant
Zygoty	Heterozygous
Chr: Position	22: 42457056
dbSNP	rs121434529
gnomAD_Freq	0.0025
gnomAD_AMR	0.00090329
gnomAD_NFE	0.00398835
gnomAD_AFR	0.00048085
gnomAD_EAS	0.00000000
gnomAD_FIN	0.00143609
gnomAD_Other	0.00318471
gnomAD_SAS	0.00251519
gnomAD_ASJ	0.00009643
gnomAD_Hom	0
Provean	-3.28 (D)
ClinVar	Conflicting interpretations of pathogenicity

This supplement provides evidence to support the classification of each reportable variant in the attached result report. This information is provided as a resource. It is not inclusive of all available information used by GeneDx for variant classification, and individual data elements may be weighted differently to derive at the classification. This information is subject to change and may differ from what is currently available. Results should always be interpreted in the context of the patient's clinical presentation. Blank fields indicate that no data were available at time of analysis.

- dbSNP - NCBI repository for single base nucleotide substitutions and short deletion and insertion polymorphisms <https://www.ncbi.nlm.nih.gov/snp/>
- The Genome Aggregation Database (gnomAD) combines exome and genome sequencing data from a variety of large-scale sequencing projects, including approximately 15,000 genomes and 123,000 exomes, including individuals recruited for disease-specific studies such as cancer and cardiovascular diseases. (PMID 32461654).
- gnomAD_Freq - variant allele frequency (in percent) from approximately 15,000 genomes and 123,000 exomes in the Genome Aggregation Database. Select ancestries include: gnomAD_AMR (Admixed American/Latino); gnomAD_AFR (African); gnomAD_EAS (East Asian); gnomAD_FIN (Finnish of European ancestry); gnomAD_NFE (non-Finnish of European ancestry); gnomAD_SAS (South Asian); gnomAD_ASJ (Ashkenazi Jewish). gnomAD_Hom - number of individuals homozygous for the variant.
- gnomAD_AMR- variant frequency (in percent) for individuals of Latino ancestry
- PROVEAN (Protein Variation Effect Analyzer) - predicts whether an amino acid substitution or indel affects the biological function of a protein using a delta alignment score from -14 to +14 (< or = -2.5, predicted deleterious; >-2.5, predicted neutral).
- Other published in silico algorithms, including those that predict splicing impact, may be considered for variant analysis. In silico scores may change. In silico models use algorithms that predict the effect a variant may have on the protein. Thus, predictions should be interpreted with caution and only be used in combination with other available evidence to support the classification of any variant (PMID 23056405).
- ClinVar - Classification of variant in ClinVar database, an NCBI archive of human variants with supporting evidence of phenotypic association. <https://www.ncbi.nlm.nih.gov/clinvar/> (PMID 26582918).

Resources

These clinician and family resources are of an informational nature only. While GeneDx makes every effort to ensure its accuracy, this information is not meant to endorse a particular group, be a complete list, nor serve as medical advice. GeneDx presents this opportunity as a courtesy to clinicians and families who may wish to obtain more details. All GeneDx genetic test reports include this Resource page, no matter the positive or negative result.

- **General support and advocacy**

- Child Neurology Foundation
www.ChildNeurologyFoundation.org
(859) 551-4977
- Global Genes
www.GlobalGenes.org
(949) 248-7273
- National Organization for Rare Disorders
www.RareDiseases.org
(617) 249-7300

- **Information exchange and connection**

- GenomeConnect: www.GenomeConnect.org
- MyGene2: www.MyGene2.org

Client/Sending Facility:
Phoenix Sperm Bank

1492 S Mill Ave Suite 306
Tempe, AZ 85281
Ph: (602)888-7255
AZB-45

LCLS Specimen Number: 263-610-3292-0

Patient Name: 10407, DONOR

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: YU19-79681 L

Indications: NOT GIVEN

[REDACTED]
Ordering Physician: J OLLIFFE

Specimen Type: BLOOD

Client Reference:

Date Collected: 09/20/2019

Date Received: 09/22/2019

Date Reported: 10/15/2019

Test: Chromosome, Blood, Routine

Cells Counted: 20

Cells Analyzed: 20

Cells Karyotyped: 2

Band Resolution: 500

CYTOGENETIC RESULT: 46,XY

INTERPRETATION: NORMAL MALE KARYOTYPE

Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTG banding pattern in all cells observed.

This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.

Chromosome analysis performed by Laboratory Corporation of America Inc., CLIA 29D0539775. 1015
Telegraph St. Suite B, Reno, NV 89502. Laboratory Director, Roger S Ritzlin, M.D.

LCLS Specimen Number: 263-610-3292-0

Patient Name: 10407, DONOR

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: YU19-79681 L

Account Number: [REDACTED]

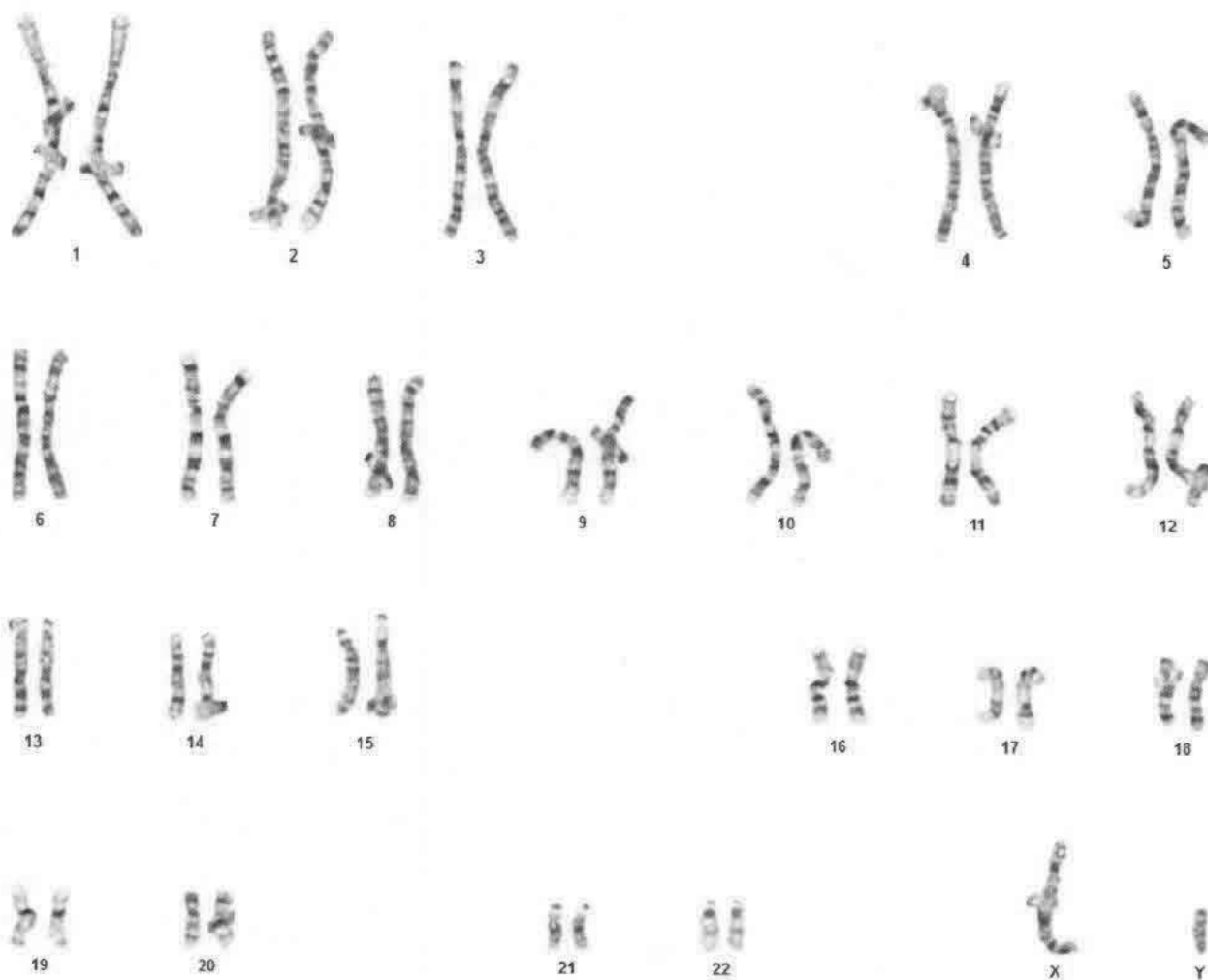
Ordering Physician: JOLLIFFE

Specimen Type: BLOOD

Client Reference:

Date Collected: 09/20/2019

Date Received: 09/22/2019





Client/Sending Facility:
Phoenix Sperm Bank

1492 S Mill Ave Suite 306
Tempe, AZ 85281
Ph: (602)888-7255
AZB-45

LCLS Specimen Number: 263-610-3292-0

Patient Name: 10407, DONOR

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: YU19-79681 L

Account Number: [REDACTED]

Ordering Physician: J OLLIFFE

Specimen Type: BLOOD

Client Reference:

Date Collected: 09/20/2019

Date Received: 09/22/2019

I. K. Gadi

Inder K. Gadi, PhD, FACMG

Arundhati Chatterjee, MD

Medical Director

Stuart Schwartz, PhD

National Director of Cytogenetics

Technical component performed by Laboratory Corporation of America Holdings,
1904 TW Alexander Drive, RTP, NC, 27709-0153 (800) 345-4363

Professional Component performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr, Research Triangle Park, NC 27709. Medical Director, Arundhati Chatterjee, MD.
Integrated Genetics is a brand used by Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

This document contains private and confidential health information protected by state and federal law.

Specimen ID: 263-610-3290-0
Control ID: B0094386918

Acct #: 02003540

Phone: (602) 888-7255

Rte: 00

10407, DONOR

Phoenix Sperm Bank
 1492 S Mill Ave Suite 306
 Tempe AZ 85281



Patient Details

DOB: [REDACTED]
Age(y/m/d): [REDACTED]
Gender: M **SSN:** [REDACTED]
Patient ID: [REDACTED]

Specimen Details

Date collected: 09/20/2019 0915 Local
Date received: 09/21/2019
Date entered: 09/21/2019
Date reported: 09/24/2019 1607 ET

Physician Details

Ordering: J OLLIFFE
Referring:
ID:
NPI: 1306838271

General Comments & Additional Information

Clinical Info: SRC: URINE

Alternate Control Number: B0094386918
Total Volume: Not Provided

Alternate Patient ID: Not Provided
Fasting: No

Ordered Items

LP+12AC+CBC/D/Plt+UA+Rh+ABO...

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
LP+12AC+CBC/D/Plt+UA+Rh+ABO...					
Chemistries					01
Glucose	97		mg/dL	65 - 99	01
Uric Acid	6.5		mg/dL	3.7 - 8.6	01
Please Note:					01
	Therapeutic target for gout patients: <6.0				
BUN	10		mg/dL	6 - 20	01
Creatinine	0.88		mg/dL	0.76 - 1.27	01
eGFR If NonAfricn Am	125		mL/min/1.73	>59	
eGFR If Africn Am	145		mL/min/1.73	>59	
Calcium	9.5		mg/dL	8.7 - 10.2	01
Protein, Total	7.2		g/dL	6.0 - 8.5	01
Albumin	4.6		g/dL	3.5 - 5.5	01
Bilirubin, Total	<0.2		mg/dL	0.0 - 1.2	01
Alkaline Phosphatase	62		IU/L	56 - 127	01
LDH	157		IU/L	121 - 224	01
AST (SGOT)	19		IU/L	0 - 40	01
ALT (SGPT)	21		IU/L	0 - 44	01
					01
Lipids					01
Cholesterol, Total	154		mg/dL	100 - 169	01
Triglycerides	59		mg/dL	0 - 89	01
HDL Cholesterol	61		mg/dL	>39	01
LDL Cholesterol Calc	81		mg/dL	0 - 109	
LDL/HDL Ratio	1.3		ratio	0.0 - 3.6	
Please Note:					01
	LDL/HDL Ratio				
				Men Women	
	1/2 Avg.Risk			1.0 1.5	
	Avg.Risk			3.6 3.2	



Patient: 10407, DONOR
DOB: [REDACTED]

Patient ID:

Control ID: B0094386918

Specimen ID: 263-610-3290-0
Date collected: 09/20/2019 0915 Local

TESTS	RESULT	FLAG	UNITS	REFERENCE	INTERVAL	LAB
			2X Avg.Risk	6.2	5.0	
			3X Avg.Risk	8.0	6.1	
Hemoglobin Fractionation						01
Hgb F	0.0		%	0.0 - 2.0		02
Hgb A	97.6		%	96.4 - 98.8		02
Hgb S	0.0		%	0.0		02
Hgb C	0.0		%	0.0		02
Hgb A2	2.4		%	1.8 - 3.2		02
Hgb Variant	0.0		%	0.0		02
Interpretation	Normal adult hemoglobin present.					02
Serology/Immunology						01
ABO Grouping	A					01
Rh Factor	Positive					01
Please note: Prior records for this patient's ABO / Rh type are not available for additional verification.						01
CBC, Platelet Ct, and Diff						01
WBC	5.3		x10E3/uL	3.4 - 10.8		01
RBC	4.92		x10E6/uL	4.14 - 5.80		01
Hemoglobin	15.4		g/dL	13.0 - 17.7		01
Hematocrit	44.6		%	37.5 - 51.0		01
MCV	91		fL	79 - 97		01
MCH	31.3		pg	26.6 - 33.0		01
MCHC	34.5		g/dL	31.5 - 35.7		01
RDW	13.9		%	12.3 - 15.4		01
Platelets	234		x10E3/uL	150 - 450		01
Neutrophils	45		%	Not Estab.		01
Lymphs	43		%	Not Estab.		01
Monocytes	10		%	Not Estab.		01
Eos	2		%	Not Estab.		01
Basos	0		%	Not Estab.		01
Neutrophils (Absolute)	2.3		x10E3/uL	1.4 - 7.0		01
Lymphs (Absolute)	2.3		x10E3/uL	0.7 - 3.1		01
Monocytes (Absolute)	0.6		x10E3/uL	0.1 - 0.9		01
Eos (Absolute)	0.1		x10E3/uL	0.0 - 0.4		01
Baso (Absolute)	0.0		x10E3/uL	0.0 - 0.2		01
Immature Granulocytes	0		%	Not Estab.		01
Immature Grans (Abs)	0.0		x10E3/uL	0.0 - 0.1		01
Urinalysis Gross Exam						01



Patient: 10407, DONOR
DOB: [REDACTED]

Patient ID:

Control ID: B0094386918

Specimen ID: 263-610-3290-0
Date collected: 09/20/2019 0915 Local

TESTS	RESULT	FLAG	UNITS	REFERENCE	INTERVAL	LAB
Specific Gravity	1.023			1.005 - 1.030		01
pH	6.0			5.0 - 7.5		01
Urine-Color	Yellow			Yellow		01
Appearance	Clear			Clear		01
WBC Esterase	Negative			Negative		01
Protein	Negative			Negative/Trace		01
Glucose	Negative			Negative		01
Ketones	Negative			Negative		01
Occult Blood	Negative			Negative		01
Bilirubin	Negative			Negative		01
Urobilinogen, Semi-Qn	0.2		mg/dL	0.2 - 1.0		01
Nitrite, Urine	Negative			Negative		01
Microscopic Examination	Microscopic not indicated and not performed.					01

01	PDLCA	LabCorp Phoenix 5005 S 40th Street Ste 1200, Phoenix, AZ 85040-2969	Dir: Earle Collum, MD
02	BN	LabCorp Burlington 1447 York Court, Burlington, NC 27215-3361	Dir: Sanjai Nagendra, MD

For inquiries, the physician may contact **Branch: 888-522-2677 Lab: 800-788-9743**

