



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Dr. Jeffrey Olliffe
4915 25th Ave NE, Suite 204W
Seattle, WA 98105
Phone: (206) 588-1484
Fax: (206) 466-4696
NPI: 1306838271
Report Date: 02/24/2017

MALE
DONOR 10123
DOB: [REDACTED]
Ethnicity: Mixed or Other
Caucasian
Sample Type: EDTA Blood
Date of Collection: 02/10/2017
Date Received: 02/12/2017
Date Tested: 02/23/2017
Barcode: 11004212111512
Indication: Egg or sperm donor

FEMALE
N/A

Family Prep Screen

POSITIVE: CARRIER

ABOUT THIS TEST

The Counsyl Family Prep Screen (version 2.0) 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 10123	Partner
Panel Information	Family Prep Screen 2.0 Universal Panel Minus X-Linked (102 conditions tested)	N/A
POSITIVE: CARRIER 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia Reproductive Risk: 1 in 230 Inheritance: Autosomal Recessive	+ CARRIER* NM_000500.7(CYP21A2):c.844G>T (V282L, aka V281L) 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.



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POSITIVE: CARRIER

21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia

Reproductive risk: 1 in 230
Risk before testing: 1 in 13,000

Gene: CYP21A2 | Inheritance Pattern: Autosomal Recessive

Patient	DONOR 10123	No partner tested
Result	⊕ Carrier	N/A
Variant(s)	NM_000500.7(CYP21A2):c.844G>T(V282L, aka V281L) heterozygote	N/A
Methodology	Analysis of homologous regions	N/A
Interpretation	This individual is a carrier of 21-hydroxylase-deficient congenital adrenal hyperplasia. Carriers generally do not experience symptoms. NM_000500.7(CYP21A2):c.844G>T is a non-classic 21-hydroxylase-deficient congenital adrenal hyperplasia mutation.	N/A
Detection rate	96%	N/A
Variants tested	CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111VfsX21, I173N, L308FfsX6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V281L, [I237N;V238E;M240K], c.293-13C>G.	N/A

What is 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia?

Congenital adrenal hyperplasia (CAH) refers to a group of genetic disorders that affect the body's adrenal glands. The adrenal glands are located above each kidney and regulate essential functions in the body, including the production of several important hormones. CAH occurs when the adrenal glands are unable to produce these hormones properly, resulting in a hormone imbalance.

More than 90% of of CAH cases are caused by deficiency of the 21-hydroxylase enzyme. When this enzyme is missing or present at low levels, the adrenal glands are unable to produce two critical hormones, cortisol and aldosterone. The body responds to this deficiency by producing an excess of male sex hormones, called androgens. Collectively, the excess androgen production and hormone deficiencies can lead to a variety of medical problems, which vary in severity depending on the form of CAH.

There are two major forms of 21-hydroxylase-deficient CAH: classic CAH and non-classic CAH.

CLASSIC

The most severe form, referred to as classic CAH, can be divided into two different subtypes: the salt-wasting type and the simple virilizing type (non salt-wasting type). The classic salt-wasting type is associated with near to complete deficiency of the enzyme, 21-hydroxylase, resulting in the complete inability to produce the hormones, cortisol and aldosterone. In this type, the body cannot retain enough sodium (salt). When too much salt is lost in the urine, it may lead to dehydration, vomiting, diarrhea, failure to thrive, heart rhythm abnormalities (arrhythmias), and shock; if not properly treated, death may occur in some cases. In addition, female newborns often have external genitalia that do not clearly appear either male or female (ambiguous genitalia), whereas male newborns may present with enlarged genitalia. Signs of early puberty (virilization) occur in both males and females with CAH. These symptoms may include: rapid growth and development in early childhood, but shorter than average height in adulthood, abnormal menstruation cycles for females, excess facial hair for females, early facial hair growth for males, severe acne, and infertility in both men and women.



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The simple virilizing type of CAH is associated with partial 21-hydroxylase deficiency. Unlike the salt-wasting type, these individuals typically do not experience severe and life-threatening sodium deficiency symptoms as newborns. However, the majority of female newborns with this type will have ambiguous genitalia, and both male and female children may show signs of early puberty.

NON-CLASSIC

The non-classic type (late-onset type) is the the least severe form of CAH and is caused by mild deficiency of the 21-hydroxylase enzyme. Individuals with this type may start experiencing symptoms related to excess androgen production in childhood, adolescence, or adulthood. Both males and females may exhibit rapid growth in childhood, shorter than average stature in adulthood, virilization, and infertility. Additionally, girls may experience symptoms of masculinization and abnormal menstruation. However, some individuals with non-classic CAH may never know they are affected because the symptoms are so mild.

How common is 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia?

The incidence of CAH varies by type and is more prevalent in certain ethnicities. Classic CAH occurs in approximately 1 in 15,000 births worldwide, while non-classic CAH is much more common, occurring in approximately 1 in 1,000 births. In some populations, namely individuals of Ashkenazi Jewish, Hispanic, Italian, and Yugoslav descent, the prevalence of the non-classic CAH can reach as high as 3-4 percent.

How is 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia treated?

Currently, there is no cure for CAH. However, treatments are available to address some of the associated symptoms. Patients benefit from taking hormone replacement medications, which work to increase levels of deficient hormones and suppress the overproduction of male hormones. Most people with classic CAH will need to take hormone medications for the rest of their life. Those with the less severe forms of CAH are sometimes able to stop taking these medications in adulthood and are typically treated with lower doses. Some individuals with non-classic CAH do not require any treatment. A multidisciplinary team of physicians, including an endocrinologist, will need to monitor the medication dosage, medication side effects, growth, and sexual development of patients who continue to receive treatment.

Newborn females with ambiguous genitalia may need surgery to correct the function and appearance of the external genitalia. Surgery, if needed, is most often performed during infancy, but can be performed later in life.

Treatments provided during pregnancy may reduce the degree of virilization in female fetuses. However, because the long term safety of prenatal treatment is unknown, these therapies are considered experimental and are not recommended by professional guidelines.

What is the prognosis for a person with 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia?

With early diagnosis and proper medication management, most individuals with CAH will have a normal life expectancy. Early death can occur during periods of significant sodium loss (salt crises) if medication dosage is not adequately adjusted, especially during times of illness or trauma. Problems with growth and development, infertility, ambiguous genitalia, and virilization are monitored by physicians on an ongoing basis.



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Methods and Limitations

DONOR 10123 [Family Prep Screen 2.0]: sequencing, targeted genotyping, copy number analysis, and analysis of homologous regions.

Sequencing

High-throughput sequencing is 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. These regions are sequenced to high coverage and the sequences are compared to standards and references of normal variation. Mutations may not be detected in areas of lower sequence coverage. On average, more than 99% of all bases in the exons listed for each gene are sequenced at the minimum read depth. Variants discovered in other exons of these genes will also be reported if they meet quality control criteria. Triplet repeats and large deletions and duplications may not be detected. Small insertions and deletions may not be as accurately determined as single nucleotide variants. Genes that have closely related pseudogenes are not well analyzed by this method.

Detection rates are calculated by estimating from literature the fraction of disease alleles that the methodology is unable to detect.

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 "predicted" or "likely" pathogenic are reported. Predicted/likely pathogenic variants are described elsewhere in the report as "predicted/likely to have a negative impact on gene function". In general, predicted pathogenic variants are those which are predicted to be pathogenic based on the nature of the sequence change, while likely pathogenic variants are evaluated by reviewing reports of allele frequencies in cases and controls, functional studies, variant annotation and effect prediction, and segregation studies. Benign variants, variants of uncertain significance, and variants not directly associated with the intended disease phenotype are not reported. Literature citations validating reported variants are available upon request.

Targeted genotyping

Targeted DNA mutation analysis is used to determine the genotypes of the listed variants in the Conditions Tested section of the report.

Copy number analysis

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. In addition, a small percentage of spinal muscular atrophy (SMA) cases are caused by nondeletion mutations in the *SMN1* gene. Thus, a test result of two *SMN1* copies significantly reduces the risk of being a carrier; however, there is still a residual risk of being a carrier and subsequently a small risk of future affected offspring for individuals with two or more *SMN1* gene copies. Some SMA cases arise as the result of *de novo* mutation events which will not be detected by carrier testing.

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.



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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. 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 Family Prep Screen 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*), and additional Tay-Sachs disease testing can be performed using a biochemical assay (*Gross et al. Genet. Med. 2008;10(1):54-56*).

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

LAB DIRECTORS

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

Conditions Tested

21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia - Gene: CYP21A2. Autosomal Recessive. Analysis of Homologous Regions. **Variants (13):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111VfsX21, I173N, L308FfsX6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V281L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Mixed or Other Caucasian 96%.

ABCC8-related Hyperinsulinism - Gene: ABCC8. Autosomal Recessive. Sequencing. **Exons:** NM_000352:1-39. **Detection Rate:** Mixed or Other Caucasian >99%.

Achromatopsia - Gene: CNGB3. Autosomal Recessive. Sequencing. **Exons:** NM_019098:1-18. **Detection Rate:** Mixed or Other Caucasian >99%.

Alkaptonuria - Gene: HGD. Autosomal Recessive. Sequencing. **Exons:** NM_000187:1-14. **Detection Rate:** Mixed or Other Caucasian >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 H5-40. **Detection Rate:** Unknown due to rarity of disease.

Alpha-1 Antitrypsin Deficiency - Gene: SERPINA1. Autosomal Recessive. Sequencing. **Exons:** NM_000295:2-5. **Detection Rate:** Mixed or Other Caucasian >99%.

Alpha-Mannosidosis - Gene: MAN2B1. Autosomal Recessive. Sequencing. **Exons:** NM_000528:1-15,17-24. **Detection Rate:** Mixed or Other Caucasian >99%.

Alpha-Sarcoglycanopathy - Gene: SGCA. Autosomal Recessive. Sequencing. **Exons:** NM_000023:1-9. **Detection Rate:** Mixed or Other Caucasian 99%.

Andermann Syndrome - Gene: SLC12A6. Autosomal Recessive. Sequencing. **Exons:** NM_133647:1-25. **Detection Rate:** Mixed or Other Caucasian >99%.

ARSACS - Gene: SACS. Autosomal Recessive. Sequencing. **Exons:** NM_014363:2-10. **Detection Rate:** Mixed or Other Caucasian 97%.

Aspartylglycosaminuria - Gene: AGA. Autosomal Recessive. Sequencing. **Exons:** NM_000027:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

Ataxia With Vitamin E Deficiency - Gene: TTPA. Autosomal Recessive. Sequencing. **Exons:** NM_000370:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.

Ataxia-Telangiectasia - Gene: ATM. Autosomal Recessive. Sequencing. **Exons:** NM_000051:2-63. **Detection Rate:** Mixed or Other Caucasian 92%.

Bardet-Biedl Syndrome, BBS1-related - Gene: BBS1. Autosomal Recessive. Sequencing. **Exons:** NM_024649:1-17. **Detection Rate:** Mixed or Other Caucasian >99%.

Bardet-Biedl Syndrome, BBS10-related - Gene: BBS10. Autosomal Recessive. Sequencing. **Exons:** NM_024685:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.

Beta-Sarcoglycanopathy - Gene: SGCB. Autosomal Recessive. Sequencing. **Exons:** NM_000232:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.

Biotinidase Deficiency - Gene: BTD. Autosomal Recessive. Sequencing. **Exons:** NM_000060:1-4. **Detection Rate:** Mixed or Other Caucasian >99%.

Bloom Syndrome - Gene: BLM. Autosomal Recessive. Sequencing. **Exons:** NM_000057:2-22. **Detection Rate:** Mixed or Other Caucasian 96%.

Canavan Disease - Gene: ASPA. Autosomal Recessive. Sequencing. **Exons:** NM_000049:1-6. **Detection Rate:** Mixed or Other Caucasian 94%.

Carnitine Palmitoyltransferase IA Deficiency - Gene: CPT1A. Autosomal Recessive. Sequencing. **Exons:** NM_001876:2-19. **Detection Rate:** Mixed or Other Caucasian 98%.

Carnitine Palmitoyltransferase II Deficiency - Gene: CPT2. Autosomal Recessive. Sequencing. **Exons:** NM_000098:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.

Cartilage-Hair Hypoplasia - Gene: RMRP. Autosomal Recessive. Sequencing. **Exon:** NR_003051:1. **Detection Rate:** Mixed or Other Caucasian >99%.

Citrullinemia Type 1 - Gene: ASS1. Autosomal Recessive. Sequencing. **Exons:** NM_000050:3-16. **Detection Rate:** Mixed or Other Caucasian >99%.

CLN3-related Neuronal Ceroid Lipofuscinosis - Gene: CLN3. Autosomal Recessive. Sequencing. **Exons:** NM_001042432:2-16. **Detection Rate:** Mixed or Other Caucasian >99%.

CLN5-related Neuronal Ceroid Lipofuscinosis - Gene: CLN5. Autosomal Recessive. Sequencing. **Exons:** NM_006493:1-4. **Detection Rate:** Mixed or Other Caucasian 98%.

Cohen Syndrome - Gene: VPS13B. Autosomal Recessive. Sequencing. **Exons:** NM_017890:2-62. **Detection Rate:** Mixed or Other Caucasian 83%.

Congenital Disorder of Glycosylation Type Ia - Gene: PMM2. Autosomal Recessive. Sequencing. **Exons:** NM_000303:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

Congenital Disorder of Glycosylation Type Ib - Gene: MPI. Autosomal Recessive. Sequencing. **Exons:** NM_002435:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

Congenital Finnish Nephrosis - Gene: NPHS1. Autosomal Recessive. Sequencing. **Exons:** NM_004646:2-23,26-27,29. **Detection Rate:** Mixed or Other Caucasian >99%.

Costeff Optic Atrophy Syndrome - Gene: OPA3. Autosomal Recessive. Sequencing. **Exons:** NM_025136:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.

Cystic Fibrosis - Gene: CFTR. Autosomal Recessive. Sequencing. **Exons:** NM_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection Rate:** Mixed or Other Caucasian 97%.

Cystinosis - Gene: CTNS. Autosomal Recessive. Sequencing. **Exons:** NM_004937:3-12. **Detection Rate:** Mixed or Other Caucasian >99%.

D-Bifunctional Protein Deficiency - Gene: HSD17B4. Autosomal Recessive. Sequencing. **Exons:** NM_000414:1-24. **Detection Rate:** Mixed or Other Caucasian 94%.

Dihydropyrimidine Dehydrogenase Deficiency - Gene: DPYD. Autosomal Recessive. Sequencing. **Exons:** NM_000110:1-23. **Detection Rate:** Mixed or Other Caucasian 93%.

Factor XI Deficiency - Gene: F11. Autosomal Recessive. Sequencing. **Exons:** NM_000128:2-15. **Detection Rate:** Mixed or Other Caucasian >99%.

Familial Dysautonomia - Gene: IKBKAP. Autosomal Recessive. Sequencing. **Exons:** NM_003640:19-20,26. **Detection Rate:** Mixed or Other Caucasian >99%.

Familial Mediterranean Fever - Gene: MEFV. Autosomal Recessive. Sequencing. **Exons:** NM_000243:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.

Fanconi Anemia Type C - Gene: FANCC. Autosomal Recessive. Sequencing. **Exons:** NM_000136:2-15. **Detection Rate:** Mixed or Other Caucasian >99%.

FKTN-related Disorders - Gene: FKTN. Autosomal Recessive. Sequencing. **Exons:** NM_001079802:3-11. **Detection Rate:** Mixed or Other Caucasian >99%.

Galactosemia - Gene: GALT. Autosomal Recessive. Sequencing. **Exons:** NM_000155:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.

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:** Mixed or Other Caucasian 60%.

GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness - Gene: GJB2. Autosomal Recessive. Sequencing. **Exons:** NM_004004:1-2. **Detection Rate:** Mixed or Other Caucasian 98%.

Glutaric Acidemia Type 1 - Gene: GCDH. Autosomal Recessive. Sequencing. **Exons:** NM_000159:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.

Glycogen Storage Disease Type Ia - Gene: G6PC. Autosomal Recessive. Sequencing. **Exons:** NM_000151:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.

Glycogen Storage Disease Type Ib - Gene: SLC37A4. Autosomal Recessive. Sequencing. **Exons:** NM_001164277:3-11. **Detection Rate:** Mixed or Other Caucasian >99%.

Glycogen Storage Disease Type III - Gene: AGL. Autosomal Recessive. Sequencing. **Exons:** NM_000642:2-34. **Detection Rate:** Mixed or Other Caucasian >99%.

Glycogen Storage Disease Type V - Gene: PYGM. Autosomal Recessive. Sequencing. **Exons:** NM_005609:1-20. **Detection Rate:** Mixed or Other Caucasian >99%.

GRACILE Syndrome - Gene: BCS1L. Autosomal Recessive. Sequencing. **Exons:** NM_004328:3-9. **Detection Rate:** Mixed or Other Caucasian >99%.

HADHA-related Disorders - Gene: HADHA. Autosomal Recessive. Sequencing. **Exons:** NM_000182:1-20. **Detection Rate:** Mixed or Other Caucasian >99%.

Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - Gene: HBB. Autosomal Recessive. Sequencing. **Exons:** NM_000518:1-3. **Detection Rate:** Mixed or Other Caucasian 96%.

Hereditary Fructose Intolerance - Gene: ALDOB. Autosomal Recessive. Sequencing. **Exons:** NM_000035:2-9. **Detection Rate:** Mixed or Other Caucasian >99%.

Herlitz Junctional Epidermolysis Bullosa, LAMA3-related - Gene: LAMA3. Autosomal Recessive. Sequencing. **Exons:** NM_000227:1-16,18-38. **Detection Rate:** Mixed or Other Caucasian >99%.



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Herlitz Junctional Epidermolysis Bullosa, LAMB3-related - Gene: LAMB3. Autosomal Recessive. Sequencing. Exons: NM_000228:2-23. **Detection Rate:** Mixed or Other Caucasian >99%.

Herlitz Junctional Epidermolysis Bullosa, LAMC2-related - Gene: LAMC2. Autosomal Recessive. Sequencing. Exons: NM_005562:1-23. **Detection Rate:** Mixed or Other Caucasian >99%.

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - Gene: HEXA. Autosomal Recessive. Sequencing. Exons: NM_000520:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency - Gene: CBS. Autosomal Recessive. Sequencing. Exons: NM_000071:3-17. **Detection Rate:** Mixed or Other Caucasian >99%.

Hypophosphatasia, Autosomal Recessive - Gene: ALPL. Autosomal Recessive. Sequencing. Exons: NM_000478:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.

Inclusion Body Myopathy 2 - Gene: GNE. Autosomal Recessive. Sequencing. Exons: NM_001128227:3-12. **Detection Rate:** Mixed or Other Caucasian >99%.

Isovaleric Acidemia - Gene: IVD. Autosomal Recessive. Sequencing. Exons: NM_002225:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.

Joubert Syndrome 2 - Gene: TMEM216. Autosomal Recessive. Sequencing. Exons: NM_001173990:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.

Krabbe Disease - Gene: GALC. Autosomal Recessive. Sequencing. Exons: NM_000153:1-17. **Detection Rate:** Mixed or Other Caucasian >99%.

Lipoamide Dehydrogenase Deficiency - Gene: DLD. Autosomal Recessive. Sequencing. Exons: NM_000108:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

Maple Syrup Urine Disease Type 1B - Gene: BCKDHB. Autosomal Recessive. Sequencing. Exons: NM_183050:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.

Medium Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADM. Autosomal Recessive. Sequencing. Exons: NM_000016:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.

Megalencephalic Leukoencephalopathy With Subcortical Cysts - Gene: MLC1. Autosomal Recessive. Sequencing. Exons: NM_015166:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.

Metachromatic Leukodystrophy - Gene: ARSA. Autosomal Recessive. Sequencing. Exons: NM_000487:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

Mucopolidosis IV - Gene: MCOLN1. Autosomal Recessive. Sequencing. Exons: NM_020533:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

Mucopolysaccharidosis Type I - Gene: IDUA. Autosomal Recessive. Targeted Genotyping. Variants (2): Q70*, W402*. **Detection Rate:** Mixed or Other Caucasian 67%.

Muscle-Eye-Brain Disease - Gene: POMGNT1. Autosomal Recessive. Sequencing. Exons: NM_017739:2-22. **Detection Rate:** Mixed or Other Caucasian 90%.

NEB-related Nemaline Myopathy - Gene: NEB. Autosomal Recessive. Sequencing. Exons: NM_004543:7-8,18,25,28,33,36,45,48,54-55,58,61,71,73-74,91,94,101,111-112,114,118-119,122-123,127,129,132-135,138,140,143,146-147. **Detection Rate:** Mixed or Other Caucasian 97%.

Niemann-Pick Disease Type C - Gene: NPC1. Autosomal Recessive. Sequencing. Exons: NM_000271:1-25. **Detection Rate:** Mixed or Other Caucasian 96%.

Niemann-Pick Disease, SMPD1-associated - Gene: SMPD1. Autosomal Recessive. Sequencing. Exons: NM_000543:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.

Nijmegen Breakage Syndrome - Gene: NBN. Autosomal Recessive. Sequencing. Exons: NM_002485:1-16. **Detection Rate:** Mixed or Other Caucasian >99%.

Northern Epilepsy - Gene: CLN8. Autosomal Recessive. Sequencing. Exons: NM_018941:2-3. **Detection Rate:** Mixed or Other Caucasian >99%.

PCDH15-related Disorders - Gene: PCDH15. Autosomal Recessive. Sequencing. Exons: NM_033056:2-33. **Detection Rate:** Mixed or Other Caucasian 85%.

Pendred Syndrome - Gene: SLC26A4. Autosomal Recessive. Sequencing. Exons: NM_000441:2-21. **Detection Rate:** Mixed or Other Caucasian >99%.

PEX1-related Zellweger Syndrome Spectrum - Gene: PEX1. Autosomal Recessive. Sequencing. Exons: NM_000466:1-24. **Detection Rate:** Mixed or Other Caucasian >99%.

Phenylalanine Hydroxylase Deficiency - Gene: PAH. Autosomal Recessive. Sequencing. Exons: NM_000277:1-13. **Detection Rate:** Mixed or Other Caucasian 98%.

PKHD1-related Autosomal Recessive Polycystic Kidney Disease - Gene: PKHD1. Autosomal Recessive. Sequencing. Exons: NM_138694:2-67. **Detection Rate:** Mixed or Other Caucasian 98%.

Polyglandular Autoimmune Syndrome Type 1 - Gene: AIRE. Autosomal Recessive. Sequencing. Exons: NM_000383:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

Pompe Disease - Gene: GAA. Autosomal Recessive. Sequencing. Exons: NM_000152:2-20. **Detection Rate:** Mixed or Other Caucasian 90%.

PPT1-related Neuronal Ceroid Lipofuscinosis - Gene: PPT1. Autosomal Recessive. Sequencing. Exons: NM_000310:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

Primary Carnitine Deficiency - Gene: SLC22A5. Autosomal Recessive. Sequencing. Exons: NM_003060:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.

Primary Hyperoxaluria Type 1 - Gene: AGXT. Autosomal Recessive. Sequencing. Exons: NM_000030:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.

Primary Hyperoxaluria Type 2 - Gene: GRHPR. Autosomal Recessive. Sequencing. Exons: NM_012203:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

PROP1-related Combined Pituitary Hormone Deficiency - Gene: PROP1. Autosomal Recessive. Sequencing. Exons: NM_006261:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.

Pseudocholinesterase Deficiency - Gene: BCHE. Autosomal Recessive. Sequencing. Exons: NM_000055:2-4. **Detection Rate:** Mixed or Other Caucasian >99%.

Pycnodysostosis - Gene: CTSK. Autosomal Recessive. Sequencing. Exons: NM_000396:2-8. **Detection Rate:** Mixed or Other Caucasian >99%.

Rhizomelic Chondrodysplasia Punctata Type 1 - Gene: PEX7. Autosomal Recessive. Sequencing. Exons: NM_000288:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.

Salla Disease - Gene: SLC17A5. Autosomal Recessive. Sequencing. Exons: NM_012434:1-11. **Detection Rate:** Mixed or Other Caucasian 93%.

Segawa Syndrome - Gene: TH. Autosomal Recessive. Sequencing. Exons: NM_000360:1-13. **Detection Rate:** Mixed or Other Caucasian 96%.

Short Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADS. Autosomal Recessive. Sequencing. Exons: NM_000017:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.

Sjogren-Larsson Syndrome - Gene: ALDH3A2. Autosomal Recessive. Sequencing. Exons: NM_000382:1-10. **Detection Rate:** Mixed or Other Caucasian 92%.

Smith-Lemli-Opitz Syndrome - Gene: DHCR7. Autosomal Recessive. Sequencing. Exons: NM_001360:3-9. **Detection Rate:** Mixed or Other Caucasian >99%.

Spinal Muscular Atrophy - Gene: SMN1. Autosomal Recessive. Copy Number Analysis. Variant (1): SMN1 copy number. **Detection Rate:** Mixed or Other Caucasian 95%.

Steroid-Resistant Nephrotic Syndrome - Gene: NPHS2. Autosomal Recessive. Sequencing. Exons: NM_014625:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

Sulfate Transporter-Related Osteochondrodysplasia - Gene: SLC26A2. Autosomal Recessive. Sequencing. Exons: NM_000112:2-3. **Detection Rate:** Mixed or Other Caucasian >99%.

TPP1-related Neuronal Ceroid Lipofuscinosis - Gene: TPP1. Autosomal Recessive. Sequencing. Exons: NM_000391:1-13. **Detection Rate:** Mixed or Other Caucasian >99%.

Tyrosinemia Type I - Gene: FAH. Autosomal Recessive. Sequencing. Exons: NM_000137:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

Usher Syndrome Type 3 - Gene: CLRN1. Autosomal Recessive. Sequencing. Exons: NM_174878:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.

Very Long Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADVL. Autosomal Recessive. Sequencing. Exons: NM_000018:1-20. **Detection Rate:** Mixed or Other Caucasian >99%.

Wilson Disease - Gene: ATP7B. Autosomal Recessive. Sequencing. Exons: NM_000053:1-21. **Detection Rate:** Mixed or Other Caucasian >99%.



RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Dr. Jeffrey Olliffe
NPI: 1306838271
Report Date: 02/24/2017

MALE
DONOR 10123
DOB: [REDACTED]
Ethnicity: Mixed or Other
Caucasian
Barcode: 11004212111512

FEMALE
N/A

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 10123 Residual Risk	Reproductive Risk
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	NM_000500.7(CYP21A2):c.844G>T(V282L, aka V281L) heterozygote †	1 in 230
ABCC8-related Hyperinsulinism	1 in 11,000	< 1 in 1,000,000
Achromatopsia	1 in 8,600	< 1 in 1,000,000
Alkaptonuria	< 1 in 50,000	< 1 in 1,000,000
Alpha Thalassemia	Alpha globin status: aa/aa.	Not calculated
Alpha-1 Antitrypsin Deficiency	1 in 3,400	1 in 460,000
Alpha-Mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-Sarcoglycanopathy	1 in 31,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 1,000,000
ARSACS	< 1 in 18,000	< 1 in 1,000,000
Aspartylglycosaminuria	< 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 2,100	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 16,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 16,000	< 1 in 1,000,000
Beta-Sarcoglycanopathy	< 1 in 50,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 12,000	< 1 in 1,000,000
Bloom Syndrome	< 1 in 12,000	< 1 in 1,000,000
Canavan Disease	< 1 in 7,700	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 31,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	< 1 in 50,000	< 1 in 1,000,000
Cartilage-Hair Hypoplasia	< 1 in 50,000	< 1 in 1,000,000
Citrullinemia Type 1	1 in 12,000	< 1 in 1,000,000
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 22,000	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 23,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 3,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ia	1 in 16,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ib	< 1 in 50,000	< 1 in 1,000,000
Congenital Finnish Nephrosis	< 1 in 50,000	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 910	1 in 99,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
D-Bifunctional Protein Deficiency	1 in 2,900	< 1 in 1,000,000
Dihydropyrimidine Dehydrogenase Deficiency	1 in 1,400	1 in 570,000
Factor XI Deficiency	< 1 in 50,000	< 1 in 1,000,000
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 Type C	1 in 16,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gaucher Disease	1 in 280	1 in 120,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 1,700	1 in 220,000
Glutaric Acidemia Type 1	1 in 10,000	< 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
Glycogen Storage Disease Type V	1 in 16,000	< 1 in 1,000,000



RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Dr. Jeffrey Olliffe
 NPI: 1306838271
 Report Date: 02/24/2017

MALE
DONOR 10123
 DOB: [REDACTED]
 Ethnicity: Mixed or Other
 Caucasian
 Barcode: 11004212111512

FEMALE
 N/A

Disease	DONOR 10123 Residual Risk	Reproductive Risk
GRACILE Syndrome	< 1 in 50,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 15,000	< 1 in 1,000,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 1,200	1 in 240,000
Hereditary Fructose Intolerance	1 in 8,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related	< 1 in 50,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-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
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency	1 in 25,000	< 1 in 1,000,000
Hypophosphatasia, Autosomal Recessive	1 in 16,000	< 1 in 1,000,000
Inclusion Body Myopathy 2	< 1 in 50,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 25,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 15,000	< 1 in 1,000,000
Lipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type 1B	1 in 25,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 5,900	< 1 in 1,000,000
Megalencephalic Leukoencephalopathy With Subcortical Cysts	< 1 in 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 20,000	< 1 in 1,000,000
Mucopolidosis IV	< 1 in 50,000	< 1 in 1,000,000
Mucopolysaccharidosis Type I	1 in 480	1 in 300,000
Muscle-Eye-Brain Disease	< 1 in 5,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	< 1 in 18,000	< 1 in 1,000,000
Niemann-Pick Disease Type C	1 in 5,400	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-associated	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	1 in 16,000	< 1 in 1,000,000
Northern Epilepsy	< 1 in 50,000	< 1 in 1,000,000
PCDH15-related Disorders	1 in 2,300	< 1 in 1,000,000
Pendred Syndrome	1 in 7,000	< 1 in 1,000,000
PEX1-related Zellweger Syndrome Spectrum	1 in 11,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 3,000	1 in 600,000
PKHD1-related Autosomal Recessive Polycystic Kidney Disease	1 in 4,100	1 in 990,000
Polyglandular Autoimmune Syndrome Type 1	1 in 14,000	< 1 in 1,000,000
Pompe Disease	1 in 1,600	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
Primary Carnitine Deficiency	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 35,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
PROP1-related Combined Pituitary Hormone Deficiency	1 in 11,000	< 1 in 1,000,000
Pseudocholinesterase Deficiency	1 in 2,700	1 in 300,000
Pycnodysostosis	< 1 in 50,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	< 1 in 1,000,000
Salla Disease	< 1 in 7,500	< 1 in 1,000,000
Segawa Syndrome	< 1 in 13,000	< 1 in 1,000,000
Short Chain Acyl-CoA Dehydrogenase Deficiency	1 in 16,000	< 1 in 1,000,000
Sjogren-Larsson Syndrome	1 in 3,100	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 4,900	1 in 970,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000
Steroid-Resistant Nephrotic Syndrome	1 in 40,000	< 1 in 1,000,000
Sulfate Transporter-Related Osteochondrodysplasia	1 in 11,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 17,000	< 1 in 1,000,000
Usher Syndrome Type 3	< 1 in 50,000	< 1 in 1,000,000
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	1 in 8,800	< 1 in 1,000,000
Wilson Disease	1 in 8,600	< 1 in 1,000,000



Patient Information:
10123, Donor
DOB: [REDACTED]
Sex: M
MR#:
Patient#: FT-PT9511811

Accession:
FT-7468009
Test#: FT-TS15636676
Order#: FT-OR15605335
Ext Test#: WP-TTS19703712
Ext Order#: WP-TOR19686097
Specimen Type: Semen
Collected: Apr 21, 2017
Received Date: Jul 10, 2026
Authorized Date: Jul 13, 2026

Physician:
[REDACTED]
Phoenix Sperm Bank
4915 25th Avenue Northeast
Ste 204
Seattle, 98105
Phone: 3039705897
Fax:

Laboratory:
Fulgent Therapeutics LLC
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Amar Jariwala
Report Date: **Jul 24, 2026**

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
<i>KCNQ1</i>	Autosomal Dominant & Autosomal Recessive	NM_000218.2:c.1343dup (p.Glu449Argfs*14)	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>)



INTERPRETATION:

ABOUT *KCNQ1*

Autosomal dominant mutations of *KCNQ1* are associated with autosomal dominant atrial fibrillation-3 (ATFB3), long QT syndrome 1 (LQTS1), short QT syndrome 2 (SQTS2); biallelic mutations of *KCNQ1* are associated with Jervell and Lange-Nielsen syndrome (JLNS). Some parent-of origin effects have been reported in this gene (PubMed: [20301579](#), [20301308](#), [17192539](#), [9020829](#); OMIM: [607554](#), [192500](#), [609621](#), [220400](#)).

***KCNQ1* NM_000218.2:c.1343dup (p.Glu449Argfs*14)**

DETECTED - Classification: Pathogenic

Targeted testing of NM_000218.2:c.1343dup (p.Glu449Argfs*14), in the *KCNQ1* 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: [rs1389696445](#). This variant is predicted to result in a frame-shift in exon 10 of the *KCNQ1* gene. This variant is predicted to introduce a premature stop codon at least 50 nucleotides upstream of the canonical donor splice site of the penultimate exon and to result in the loss of function of the protein product due to nonsense-mediated mRNA decay (PubMed: [25741868](#), [30192042](#), [27618451](#), [11532962](#), [18066079](#)). Loss-of-function variants in the *KCNQ1* gene have been associated with Long QT syndrome 1 (PubMed: [23098067](#), [26669661](#), [29532034](#)). A frameshift variant downstream of this alteration has been reported to be pathogenic (PubMed: [30942114](#)). This variant, also known as 1338insC, has been reported in multiple related individuals with long QT syndrome (PubMed: [12702160](#)). This variant has also been reported in the homozygous state in an individual with Jervell and Lange-Nielsen syndrome (PubMed: [28364778](#)). This variant is classified as a "Disease Mutation" (DM) in the Human Gene Mutation Database (HGMD). This variant has one or more entries in ClinVar: RCV000045982.5, RCV000182282.3. This variant has been observed at a frequency of less than 0.01% (1/250342 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 less than 0.01% in the Latino 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.



GENES TESTED:

Known Mutation / Site-Specific Testing

1 genes tested

KCNQ1

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:



Dr. Harry Gao, DABMG, FACMG on 07/24/2026
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.

Client/Sending Facility:
Phoenix Sperm Bank

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

LCLS Specimen Number: 041-944-3178-0

Patient Name: 10123, DONOR

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: YU17-11706 L

Indications: NOT GIVEN

Account Number: [REDACTED]

Ordering Physician: J OLLIFFE

Specimen Type: BLOOD

Client Reference:

Date Collected: 02/10/2017

Date Received: 02/12/2017

Date Reported: 02/22/2017

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 Esoterix Genetics Laboratories, LLC, CLIA 32D0653253, 2000 Vivigen Way, Santa Fe, NM 87505.



Client/Sending Facility:
Phoenix Sperm Bank

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

LCLS Specimen Number: 041-944-3178-0

Patient Name: 10123, DONOR

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: YU17-11706 L

Account Number: [REDACTED]

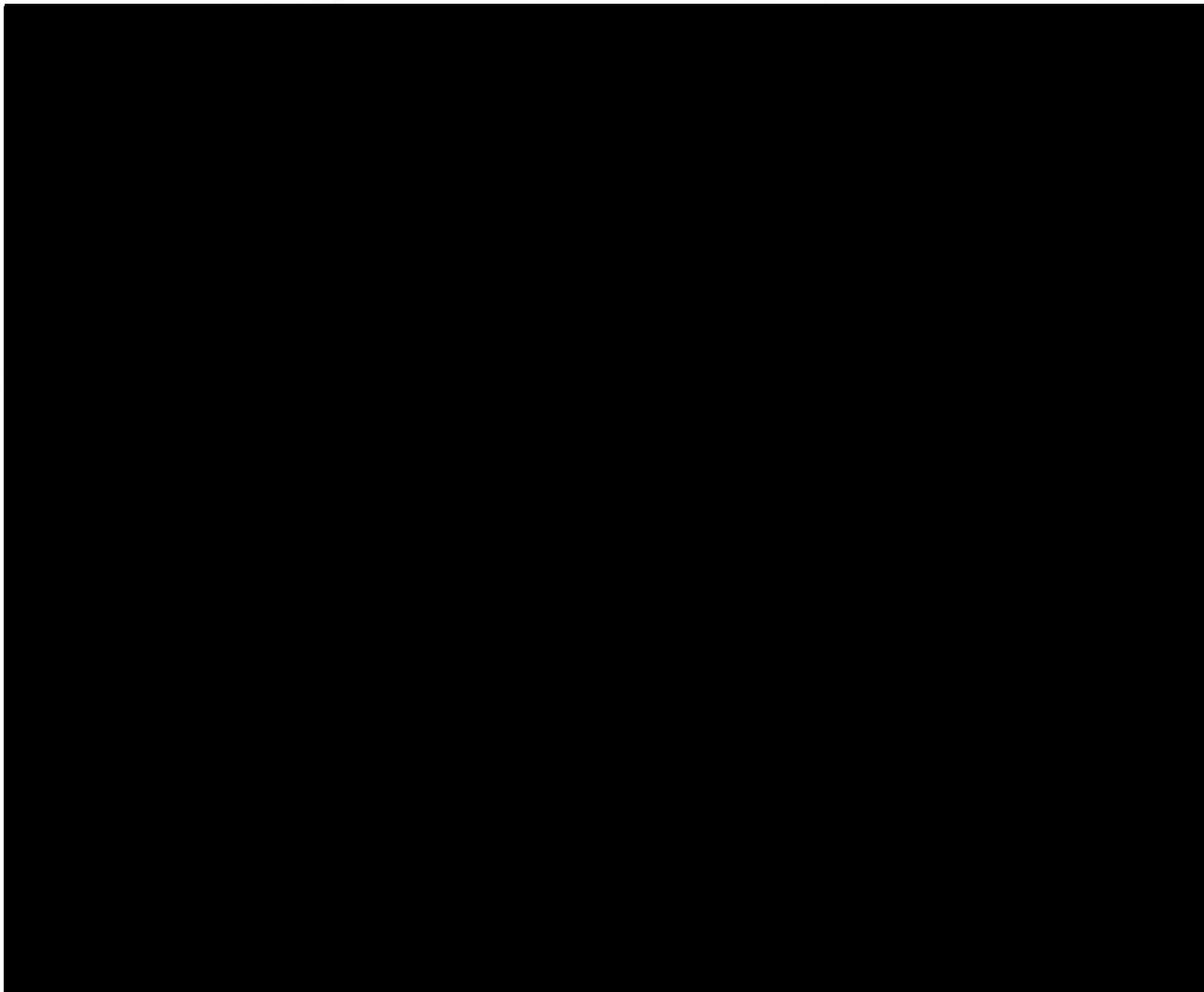
Ordering Physician: J OLLIFFE

Specimen Type: BLOOD

Client Reference:

Date Collected: 02/10/2017

Date Received: 02/12/2017





Client/Sending Facility:
Phoenix Sperm Bank

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

LCLS Specimen Number: 041-944-3178-0

Patient Name: 10123, DONOR

Date of Birth: [REDACTED]

Gender: M

Patient ID:

Lab Number: YU17-11706 L

Account Number: [REDACTED]

Ordering Physician: J OLLIFFE

Specimen Type: BLOOD

Client Reference:

Date Collected: 02/10/2017

Date Received: 02/12/2017

James H. Tepperberg, PhD, FACMG
Board Certified Cytogeneticist

Arundhati Chatterjee, MD
Medical Director
Peter Papenhausen, 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.**

Patient: 10123, DONOR
DOB: [REDACTED]

Patient ID:

Control ID: 80054708398

Specimen ID: 041-944-3170-0
Date collected: 02/10/2017 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	97.5		%	94.0 - 98.0		02
Hgb S	0.0		%	0.0		02
Hgb C	0.0		%	0.0		02
Hgb A2	2.5		%	0.7 - 3.1		02
Interpretation	Normal adult hemoglobin present.					02
Serology/Immunology						01
ABO Grouping	O					01
Rh Factor	Positive					01
Please note: Prior records for this patient's ABO / Rh type are not available for additional verification.						
CBC, Platelet Ct, and Diff						01
WBC	10.6		x10E3/uL	3.4 - 10.8		01
RBC	4.92		x10E6/uL	4.14 - 5.80		01
Hemoglobin	16.2		g/dL	12.6 - 17.7		01
Hematocrit	47.3		%	37.5 - 51.0		01
MCV	96		fL	79 - 97		01
MCH	32.9		pg	26.6 - 33.0		01
MCHC	34.2		g/dL	31.5 - 35.7		01
RDW	13.1		%	12.3 - 15.4		01
Platelets	281		x10E3/uL	150 - 379		01
Neutrophils	70		%			01
Lymphs	23		%			01
Monocytes	6		%			01
Eos	1		%			01
Basos	0		%			01
Neutrophils (Absolute)	7.3	High	x10E3/uL	1.4 - 7.0		01
Lymphs (Absolute)	2.5		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		%			01
Immature Grans (Abs)	0.0		x10E3/uL	0.0 - 0.1		01
Urinalysis Gross Exam						01
Specific Gravity	1.005			1.005 - 1.030		01

[Handwritten signature] 10 Feb 17

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FINAL REPORT

Page 2 of 3

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