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Patient Information:

13144, DONOR

DOB: [REDACTED]

Sex: M

MR#:

Patient#: FT-PT8686659

Partner Information:

Not Tested

Physician:

Kuan, James

ATTN: Kuan, James

Seattle Sperm Bank

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Laboratory:

Fulgent Therapeutics LLC

CAP#: 8042697

CLIA#: 05D2043189

Laboratory Director:

Dr. Hanlin (Harry) Gao

Report Date: **Apr 05, 2024**

Accession:

FT-7160554

Test#: FT-TS14793336

Specimen Type: Blood (EDTA)

Collected: Mar 20, 2024

Accession:

N/A

FINAL RESULTS



Carrier for genetic conditions in **multiple** genes.
Genetic counseling is recommended.

TEST PERFORMED

Beacon Preconception Carrier Screening - 515 Genes (without X-linked Disorders)

(515 Gene Panel; gene sequencing with deletion and duplication analysis)

Condition and Gene	Inheritance	13144, DONOR	Partner
Carnitine palmitoyltransferase 1A deficiency <i>CPT1A</i>	AR	⊕ Carrier c.1436C>T (p.Pro479Leu)	N/A
Smith-Lemli-Opitz syndrome <i>DHCR7</i>	AR	⊕ Carrier c.964-1G>C (p.?)	N/A
Congenital disorder of glycosylation type 1k <i>ALG1</i>	AR	⊕ Carrier c.989del (p.Ser330Thrfs*28)	N/A

INTERPRETATION:

Notes and Recommendations:

- Based on these results, this individual is positive for carrier mutations in 3 genes. Carrier screening for the reproductive partner is recommended to accurately assess the risk for any autosomal recessive conditions. A negative result reduces, but does not eliminate, the chance to be a carrier for any condition included in this screen. Please see the supplemental table for details.
- Testing for copy number changes in the SMN1 gene was performed to screen for the carrier status of Spinal Muscular Atrophy. The results for this individual are within the normal range for non-carriers. See Limitations section for more information.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. This report does not include variants of uncertain significance; only variants classified as pathogenic or likely pathogenic at the time of testing, and considered relevant for reproductive carrier screening, are reported. Please see the gene specific notes for details. Please note that the classification of variants can change over time.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- X-linked genes are not routinely analyzed for male carrier screening tests. Gene specific notes and limitations may be present. See below.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; <https://www.nsgc.org>).

Patient: 13144, DONOR; Sex: M;
DOB: [REDACTED]; **MR#:**

Accession#: FT-7160554; FD Patient#: FT-PT8686659;
DocID: FT-TS14793336AA; **PAGE 1 of 7**



CARNITINE PALMITOYLTRANSFERASE 1A DEFICIENCY

Patient	13144, DONOR	Partner
Result	 Carrier	N/A
Variant Details	CPT1A (NM_001876.4) c.1436C>T (p.Pro479Leu)	N/A

What is Carnitine palmitoyltransferase 1A deficiency?

Carnitine palmitoyltransferase 1A (CPT-1A) deficiency manifests between birth and 18 months of age with recurrent attacks of hypoketotic hypoglycemia of varying severity, triggered by fasting or illness. These episodes of hypoglycemia can lead to neurological disease. Patients may also present with hepatic encephalopathy, loss of consciousness, seizures, coma, or even sudden death. There may be a risk of progression to liver failure. Some patients with severe CPT-1A deficiency may also have renal tubular acidosis.

What is my risk of having an affected child?

Carnitine palmitoyltransferase 1A deficiency is inherited in an autosomal recessive manner. If the patient and the partner are both carriers, the risk for an affected child is 1 in 4 (25%).

What kind of medical management is available?

Treatment consists primarily of avoidance of fasting and specialized dietary supplementation. Regular surveillance of liver enzymes and function is necessary. With treatment, the prognosis is good if episodes of recurrent hypoglycemia are prevented.

What mutation was detected?

The detected heterozygous variant was NM_001876.4:c.1436C>T (p.Pro479Leu). This variant has been reported in the homozygous state in several individuals with hypoketotic hypoglycemia and/ or infant mortality (PubMed: [11441142](#), [26820065](#), [25449608](#), [28125087](#)). This variant has been found in high frequency (allele frequency ranging from 0.68 to 0.85) in Arctic populations (such as Alaska, Canada, Greenland, and Northeast Siberia (PubMed: [26820065](#)). Experimental studies in cultured fibroblasts and COS cells have demonstrated that this variant results in partially impaired (~20% of normal) L-CPTI activity, with a diminished response to malonyl-CoA (PubMed: [11441142](#), [27341449](#)). This mutation may have conferred a metabolic advantage for the Northeast Siberian populations in dealing with their traditional high-fat diet, thereby exerting a cardioprotective role through its association with elevated levels of high-density lipoprotein cholesterol and reduced adiposity. However, homozygous children for this variant have repeatedly been shown to have significantly impaired fasting ketogenesis resulting in hypoketotic hypoglycemia, liver dysfunction, and sudden infant death (PubMed: [21763168](#), [32561900](#)). The laboratory classifies this variant as pathogenic.



SMITH-LEMLI-OPITZ SYNDROME

Patient	13144, DONOR	Partner
Result	 Carrier	N/A
Variant Details	DHCR7 (NM_001360.3) c.964-1G>C (p.?)	N/A

What is Smith-Lemli-Opitz syndrome?

Smith-Lemli-Opitz syndrome (SLOS) is an impairment of the body's ability to make cholesterol. As a result, affected infants typically have cleft palate, hypotonia (low muscle syndrome), and failure to thrive. Other characteristics can include ambiguous genitalia, cardiac defects, kidney problems, dysmorphic facial features, microcephaly (abnormally small head), syndactyly (webbed or conjoined fingers or toes), and intellectual disability. The clinical features and severity of symptoms vary from person to person.

What is my risk of having an affected child?

SLOS is inherited in an autosomal recessive manner. This means that when both parents are carriers for the condition, there is a 25% (1 in 4) risk of having an affected child.

What kind of medical management is available?

There is currently no cure for SLOS, but symptoms can be addressed. Dietary cholesterol supplementation may improve clinical symptoms. Additionally, early intervention with physical, speech, and occupational therapy may improve developmental issues.

What mutation was detected?

The detected heterozygous variant was NM_001360.3:c.964-1G>C (p.?). This intronic variant, c.964-1G>C, alters the highly conserved splice acceptor site for exon 9 of this transcript and is predicted by all four splice site prediction tools queried to abolish the canonical splice acceptor site. A functional study in patient-derived mRNA demonstrated that this variant disrupts the canonical acceptor site, resulting in the use of a cryptic splice acceptor site, the insertion of 134 bp, a frameshift, and a premature stop codon (PubMed: [9653161](#)). There's sufficient evidence that loss of function in this gene is a known disease mechanism for Smith-Lemli-Opitz syndrome (PubMed: [11111101](#), [16618793](#), [22226660](#)). This variant accounts for a third of all mutant alleles in the **DHCR7** gene in individuals with Smith-Lemli-Opitz syndrome (SLOS) (PubMed: [9653161](#), [10995508](#), [12949967](#), [17965227](#), [29455191](#), [28805615](#), [22226660](#), [28369852](#), [12818773](#), [12914579](#), [22929031](#)). This variant is classified as "Pathogenic" or "Likely Pathogenic" in ClinVar, with multiple submitters in agreement (ClinVar: 93725). The laboratory classifies this variant as pathogenic.



CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1K

Patient	13144, DONOR	Partner
Result	 Carrier	N/A
Variant Details	ALG1 (NM_019109.5) c.989del (p.Ser330Thrfs*28)	N/A

What is Congenital disorder of glycosylation type 1k?

Congenital disorder of glycosylation, type 1k is an inherited metabolic disease with varying signs and symptoms that typically develop during infancy and can affect several body systems. The phenotypic spectrum of this condition ranges from mild intellectual disability to death in the first few weeks of life. Features may include severe developmental delay, rapidly progressive microcephaly, hypotonia, early-onset seizures, severe coagulation defects, immunodeficiency, nephrotic syndrome, liver dysfunction, and cardiomyopathy.

What is my risk of having an affected child?

Congenital disorder of glycosylation, type 1k is inherited in an autosomal recessive manner. If the patient and the partner are both carriers, the risk for an affected child is 1 in 4 (25%).

What kind of medical management is available?

There is no specific treatment for the disorder itself. Instead, overall treatment is mostly focused on the specific symptoms that which can vary from patient to patient, often requiring multiple medical specialties. Occupational, physical, and speech therapies may also be helpful.

What mutation was detected?

The detected heterozygous variant was NM_019109.5:c.989del (p.Ser330Thrfs*28). 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](#)). There's sufficient evidence that loss of function in this gene is a known disease mechanism for congenital disorder of glycosylation type 1k (CDG-1k) (PubMed: [26931382](#), [30653653](#)). The laboratory classifies this variant as likely pathogenic.



GENES TESTED:

Beacon Preconception Carrier Screening - 515 Genes (without X-linked Disorders) - 515 Genes

This analysis was run using the Beacon Preconception Carrier Screening - 515 Genes (without X-linked Disorders) gene list. 515 genes were tested with 99.5% of targets sequenced at >20x coverage. For more gene-specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

AAAS, ABCA12, ABCA3, ABCA4, ABCB11, ABCB4, ABCC2, ABCC8, ACAD9, ACADM, ACADVL, ACAT1, ACOX1, ACSF3, ADA, ADAMTS2, ADAMTSL4, ADGRG1, ADGRV1, AGA, AGL, AGPS, AGXT, AHI1, AIPL1, AIRE, ALDH3A2, ALDH7A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, AMN, AMT, ANO10, AP1S1, AQP2, ARG1, ARL6, ARSA, ARSB, ASL, ASNS, ASPA, ASS1, ATM, ATP6V1B1, ATP7B, ATP8B1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCKDHA, BCKDHB, BCS1L, BLM, BLOC1S3, BLOC1S6, BMP1, BRIP1, BSND, CAD, CANT1, CAPN3, CASQ2, CBS, CC2D1A, CC2D2A, CCDC103, CCDC39, CCDC88C, CD3D, CD3E, CD40, CD59, CDH23, CEP152, CEP290, CERKL, CFTR, CHAT, CHRNE, CHRNA, CIITA, CLCN1, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COL11A2, COL17A1, COL27A1, COL4A3, COL4A4, COL7A1, COX15, CPS1, CPT1A, CPT2, CRB1, CRTAP, CRYL1, CTNS, CTSA, CTSC, CTSD, CTSK, CYBA, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP21A2, CYP27A1, CYP27B1, CYP7B1, DBT, DCAF17, DCLRE1C, DDX11, DGAT1, DGUOK, DHCR7, DHDDS, DLD, DLL3, DNAH11, DNAH5, DNAI1, DNAI2, DNMT3B, DOK7, DUOX2, DYNC2H1, DYSF, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELP1, EPG5, ERCC2, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYS, FAH, FAM161A, FANCA, FANCC, FANCD2, FANCE, FANCG, FANCI, FANCL, FBP1, FBXO7, FH, FKBP10, FKBP, FKTN, FMO3, FOXN1, FOXRED1, FRAS1, FREM2, FUCA1, G6PC, G6PC3, GAA, GALT, GALE, GALK1, GALNS, GALNT3, GALT, GAMT, GATM, GBA, GBE1, GCDH, GCH1, GDF5, GFM1, GHR, GJB2, GJB6, GJB1, GLDC, GLE1, GNE, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GRHR, GRIP1, GSS, GUCY2D, GUSB, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HEXA, HEXB, HGSNAT, HJV, HLCS, HMGCL, HMOX1, HOGA1, HPD, HPS1, HPS3, HPS4, HPS5, HPS6, HSD17B3, HSD17B4, HSD3B2, HYAL1, HYLS1, IDUA, IGHMBP2, IKBK, IL7R, INVS, ITGA6, ITGB3, ITGB4, IVD, JAK3, KCNJ1, KCNJ11, LAMA2, LAMA3, LAMB3, LAMC2, LARGE1, LCA5, LDLR, LDLRAP1, LHX3, LIFR, LIG4, LIPA, LMBRD1, LOXHD1, LPL, LRAT, LRP2, LRPPRC, LYST, MAK, MAN2B1, MANBA, MCEE, MCOLN1, MCPH1, MECP, MED17, MESF2, MFSDB, MKKS, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MPI, MPL, MPV17, MRE11, MTHFR, MTR, MTRR, MTPP, MUSK, MUT, MVK, MYO15A, MYO7A, NAGA, NAGLU, NAGS, NBN, NCF2, NDRG1, NDUFAF2, NDUFAF5, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NEB, NEU1, NGLY1, NPC1, NPC2, NPHP1, NPHS1, NPHS2, NR2E3, NSMCE3, NTRK1, OAT, OCA2, OPA3, OSTM1, OTOA, OTOF, P3H1, PAH, PANK2, PC, PCBD1, PCCA, PCCB, PCDH15, PCNT, PDHB, PEPD, PET100, PEX1, PEX10, PEX12, PEX13, PEX16, PEX2, PEX26, PEX5, PEX6, PEX7, PFKM, PGM3, PHGDH, PHKB, PHKG2, PHYH, PIGN, PJKV, PKHD1, PLA2G6, PLEKHG5, PLOD1, PMM2, PNPO, POLG, POLH, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PRCD, PRDM5, PRF1, PROP1, PSAP, PTPRC, PTS, PUS1, PYGM, QDPR, RAB23, RAG1, RAG2, RAPSN, RARS2, RDH12, RLBP1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RPE65, RPGRIP1L, RTEL1, RXYLT1, RYR1, SACS, SAMD9, SAMHD1, SCO2, SEC23B, SEPSecs, SGCA, SGCB, SGCD, SGCG, SGSH, SKIV2L, SLC12A1, SLC12A3, SLC12A6, SLC17A5, SLC19A2, SLC19A3, SLC1A4, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC26A2, SLC26A3, SLC26A4, SLC27A4, SLC35A3, SLC37A4, SLC38A8, SLC39A4, SLC45A2, SLC4A11, SLC5A5, SLC7A7, SMARCA1, SMN1, SMPD1, SNAP29, SPG11, SPR, SRD5A2, ST3GAL5, STAR, STX11, STXBP2, SUMF1, SUOX, SURF1, SYNE4, TANGO2, TAT, TBCD, TBCE, TCIRG1, TCN2, TECPR2, TERT, TF, TFR2, TG, TGM1, TH, TK2, TMC1, TMEM216, TMEM67, TMPPRS3, TPO, TPP1, TREX1, TRIM32, TRIM37, TRMU, TSEN54, TSFM, TSHB, TSHR, TTC37, TTPA, TULP1, TYMP, TYR, TYRP1, UBR1, UNC13D, USH1C, USH2A, VDR, VLDLR, VPS11, VPS13A, VPS13B, VPS45, VPS53, VRK1, VSX2, WISP3, WNT10A, WRN, XPA, XPC, ZBTB24, ZFYVE26, ZNF469

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). 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), variants were detected in regions of at least 10x coverage. For this specimen, 99.58% and 99.55% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

LIMITATIONS:

General Limitations

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or



otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. 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 available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

Gene Specific Notes and Limitations

ALG1: Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in exons 6-13 of the ALG1 gene (NM_019109.4). **CEP290:** Copy number analysis for exons 8-13 and exons 39-42 may have reduced sensitivity in the CEP290 gene. Confirmation of these exons are limited to individuals with a positive personal history of CEP290-related conditions and/or individuals carrying a pathogenic/likely pathogenic sequence variant. **CFTR:** Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. **CRYL1:** As mutations in the CRYL1 gene are not known to be associated with any clinical condition, sequence variants in this gene are not analyzed. However, to increase copy number detection sensitivity for large deletions including this gene and a neighboring gene on the panel (GJB6, also known as connexin 30), this gene was evaluated for copy number variation. **CYP11B1:** The current testing method is not able to reliably detect certain pathogenic variants in this gene due to the interference by highly homologous regions. This analysis is not designed to detect or rule-out copy-neutral chimeric CYP11B1/CYP11B2 gene. **CYP11B2:** The current testing method is not able to reliably detect certain pathogenic variants in this gene due to the interference by highly homologous regions. This analysis is not designed to detect or rule-out copy-neutral chimeric CYP11B1/CYP11B2 gene. **CYP21A2:** Significant pseudogene interference and/or reciprocal exchanges between the CYP21A2 gene and its pseudogene, CYP21A1P, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of this individual's clinical findings, biochemical profile, and family history. **DDX11:** Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in the DDX11 gene. **DUOX2:** The current testing method is not able to reliably detect variants in exons 6-8 of the DUOX2 gene (NM_014080.5) due to significant interference by the highly homologous gene, DUOX1. **FANCD2:** Due to pseudogene interference, copy-number-variants within exon 14-17 of the FANCD2 gene (NM_033084.4) are not evaluated and detection of single-nucleotide variants and small insertions/deletions in this region is not guaranteed. **GALT:** In general, the D2 "Duarte" allele is not reported if detected, but can be reported upon request. While this allele can cause positive newborn screening results, it is not known to cause clinical symptoms in any state. See GeneReviews for more information: <https://www.ncbi.nlm.nih.gov/books/NBK1518/>. **GBA:** Significant pseudogene interference and/or reciprocal exchanges between the GBA gene and its pseudogene, GBAP1, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of this individual's clinical findings, biochemical profile, and family history. The current testing method cannot detect copy-neutral rearrangements between the pseudogene and the functional gene, which have been reported in very rare cases of Gaucher disease (PubMed: 21704274). **HBA1:** Significant interference from highly homologous regions in exons 1-2 of the HBA1 gene has been recognized to occur, potentially impeding the assay's technical capability to detect pathogenic alterations during sequencing analyses. **HBA2:** Significant interference from highly homologous regions in exons 1-2 of the HBA2 gene has been recognized to occur, potentially impeding the assay's technical capability to detect pathogenic alterations during sequencing analyses. **HSD17B4:** Copy number analysis for exons 4-6 may have reduced sensitivity in the HSD17B4 gene. Confirmation of these exons are limited to individuals with a positive personal history of D-bifunctional protein deficiency and Perrault syndrome and/or individuals carrying a pathogenic/likely pathogenic sequence variant. **LMBRD1:** Copy number analysis for exons 9-12 may have reduced sensitivity in the LMBRD1 gene. Confirmation of these exons are limited to individuals with a positive personal history of combined



methylmalonic aciduria and homocystinuria and/or individuals carrying a pathogenic/likely pathogenic sequence variant. **MTHFR:** As recommended by ACMG, the two common polymorphisms in the MTHFR gene - c.1286A>C (p.Glu429Ala, also known as c.1298A>C) and c.665C>T (p.Ala222Val, also known as c.677C>T) - are not reported in this test due to lack of sufficient clinical utility to merit testing (PubMed: [23288205](#)). **NEB:** This gene contains a 32-kb triplicate region (exons 82-105) which is not amenable to sequencing and deletion/duplication analysis. **OTOA:** Due to pseudogene interference, our current testing method is not able to reliably detect variants in exons 20-28 (NM_144672.3) in the OTOA gene. **SMN1:** The current testing method detects sequencing variants and copy number variation in exons 7 of the SMN1 gene (NM_022874.2). Sequencing and deletion/duplication analysis are not performed on any other region in this gene. **TERT:** The TERT promoter region is analyzed for both sequencing and copy number variants. **TYR:** Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in exons 4-5 of the TYR gene (NM_000372.5). **WRN:** Due to the interference by highly homologous regions within the WRN gene, our current testing method has less sensitivity to detect variants in exons 10-11 of WRN (NM_000553.6).

SIGNATURE:



Geetu Mendiratta-Vij, PhD, FACMG, CGMBS on 4/5/2024
Laboratory Director, Fulgent

DISCLAIMER:

This test was developed 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](tel:626-350-0537) or by email at 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.

To view the supplemental table describing the carrier frequencies, detection rates, and residual risks associated with the genes on this test please visit the following link:

[Beacon Expanded Carrier Screening Supplemental Table](#)



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Lab Director: Arash Radfar M.D.
CLIA: 22D0957540



Patient Information:

13144, DONOR

DOB: [REDACTED]

Sex: M

MR#:

Patient#: FT-PT8686659

Partner Information:

Not Tested

Physician:

Kuan, James

ATTN: Kuan, James

Seattle Sperm Bank

4915 25th Avenue NE, Ste 204W

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Laboratory:

Fulgent Therapeutics LLC

CAP#: 8042697

CLIA#: 05D2043189

Laboratory Director:

Dr. Amar Jariwala

Report Date: **May 05, 2026**

Accession:

FT-7160554

Test#: FT-TS15564864

Specimen Type: Blood (EDTA)

Collected: Mar 20, 2024

Accession:

N/A

FINAL RESULTS



No carrier mutations identified

TEST PERFORMED

Custom Beacon Preconception Carrier Screening Panel

(2 Gene Panel: *BCHE* and *DPYD*; gene sequencing with deletion and duplication analysis)

INTERPRETATION:

Notes and Recommendations:

- No carrier mutations were identified in the submitted specimen. A negative result does not rule out the possibility of a genetic predisposition nor does it rule out any pathogenic mutations in areas not assessed by this test or in regions that were covered at a level too low to reliably assess. Also, it does not rule out mutations that are of the sort not queried by this test; see Methods and Limitations for more information. A negative result reduces, but does not eliminate, the chance to be a carrier for any condition included in this screen. Please see the supplemental table for details.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. This report does not include variants of uncertain significance; only variants classified as pathogenic or likely pathogenic at the time of testing, and considered relevant for reproductive carrier screening, are reported. Please see the gene specific notes for details. Please note that the classification of variants can change over time.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- Gene specific notes and limitations may be present. See below.
- 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>).



GENES TESTED:

Custom Beacon Preconception Carrier Screening Panel - 2 Genes

This analysis was run using the Custom Beacon Preconception Carrier Screening Panel gene list. 2 genes were tested with 100.0% of targets sequenced at >20x coverage. For more gene-specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

BCHE, DPYD

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). 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), variants were detected in regions of at least 10x coverage. For this specimen, 100.00% and 100.00% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The FPLMv2.0 pipeline was used to analyze this specimen.

LIMITATIONS:

General Limitations

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. 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 available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. Saliva swab samples may have reduced sensitivity for CNVs and a higher CNV failure rate. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

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Lab Director: Arash Radfar M.D.
CLIA: 22D0957540



Gene Specific Notes and Limitations

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

SIGNATURE:

A handwritten signature in black ink, appearing to read "Wei Kelly", written over a horizontal line.

Wei Kelly M.D., Ph.D., FACMG, CGMBS on 05/05/2026
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 by email at 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.

To view the supplemental table describing the carrier frequencies, detection rates, and residual risks associated with the genes tested on any Beacon panel, please visit the following link:

[Beacon Expanded Carrier Screening Supplemental Table](#)



Patient: 13144, DONOR; Sex: M;
DOB: [REDACTED] MR#:

Accession#: FT-7160554; FD Patient#: FT-PT8686659;
DocID: FT-TS15564864AA; **PAGE 3 of 3**



Patient Information	Specimen Information	Client Information
13144, DONOR DOB: [REDACTED] AGE: [REDACTED] Gender: M Fasting: U Phone: 206.588.1484 Patient ID: 13144 Health ID: 8573034249035303	Specimen: OW648690Y Requisition: 0000634 Collected: 03/20/2024 / 12:59 PDT Received: 03/21/2024 / 03:34 PDT Reported: 04/03/2024 / 20:24 PDT	Client #: 98105026 VNLZR00 KUAN, JAMES K SEATTLE SPERM BANK 4915 25TH AVE NE STE 204W SEATTLE, WA 98105-5668

COMMENTS: FASTING:UNKNOWN

Cytogenetic Report

CHROMOSOME ANALYSIS, BLOOD - 14596

Lab:EZ

CHROMOSOME ANALYSIS, BLOOD

Order ID: 24-136081
Specimen Type: Blood
Clinical Indication: GAMETE DONOR

RESULT:
NORMAL MALE KARYOTYPE

INTERPRETATION:

Chromosome analysis revealed normal G-band patterns within the limits of standard cytogenetic analysis.

Please expect the results of any other concurrent study in a separate report.

NOMENCLATURE:

46,XY

ASSAY INFORMATION:

Method: G-Band (Digital Analysis: MetaSyst)
Cells Counted: 30
Band Level: 450
Cells Analyzed: 5
Cells Karyotyped: 5

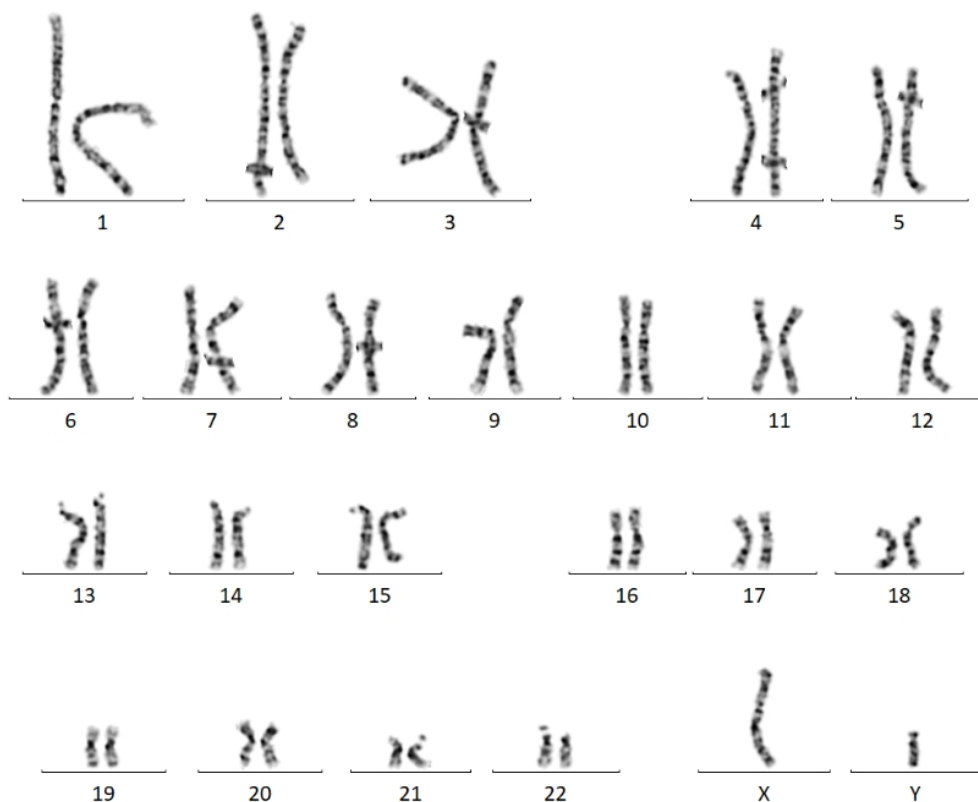
This test does not address genetic disorders that cannot be detected by standard cytogenetic methods or rare events such as low level mosaicism or subtle rearrangements.

Mark A. Micale, PhD, FACMG, [site SJC6]

Electronic Signature: 4/3/2024 10:44 PM



Patient Information	Specimen Information	Client Information
13144, DONOR DOB: [REDACTED] AGE: [REDACTED] Gender: M Fasting: U Patient ID: 13144 Health ID: 8573034249035303	Specimen: OW648690Y Collected: 03/20/2024 / 12:59 PDT Received: 03/21/2024 / 03:34 PDT Reported: 04/03/2024 / 20:24 PDT	Client #: 98105026 KUAN, JAMES K



PERFORMING SITE:

EZ QUEST DIAGNOSTICS/NICHOLS SJC, 33608 ORTEGA HWY, SAN JUAN CAPISTRANO, CA 92675-2042 Laboratory Director: IRINA MARAMICA,MD,PHD,MBA, CLIA: 05D0643352

Date Collected: 03/20/2024

Date Received: 03/21/2024

Date Reported: 03/27/2024

Fasting: Not Given

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

Date Collected: 03/20/2024

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

Test	Current Result and Flag		Previous Result and Date	Units	Reference Interval
Glucose ⁰¹	90			mg/dL	70-99
Uric Acid ⁰¹	5.9			mg/dL	3.8-8.4
	Therapeutic target for gout patients: <6.0				
BUN ⁰¹	17			mg/dL	6-20
Creatinine ⁰¹	1.13			mg/dL	0.76-1.27
eGFR	87			mL/min/1.73	>59
Calcium ⁰¹	9.2			mg/dL	8.7-10.2
Protein, Total ⁰¹	6.7			g/dL	6.0-8.5
Albumin ⁰¹	4.4			g/dL	4.1-5.1
Bilirubin, Total ⁰¹	0.2			mg/dL	0.0-1.2
Alkaline Phosphatase ⁰¹	60			IU/L	44-121
LDH ⁰¹	164			IU/L	121-224
AST (SGOT) ⁰¹	25			IU/L	0-40
ALT (SGPT) ⁰¹	29			IU/L	0-44
Cholesterol, Total ⁰¹	164			mg/dL	100-199
Triglycerides ⁰¹	95			mg/dL	0-149
▼ HDL Cholesterol ⁰¹	38	Low		mg/dL	>39
▲ LDL Chol Calc (NIH)	108	High		mg/dL	0-99
LDL/HDL Ratio	2.8			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
2X Avg.Risk		6.2	5.0
3X Avg.Risk		8.0	6.1

Hgb Fractionation by CE: ⁰²				
Hgb F ⁰²	0.0		%	0.0-2.0
Hgb A ⁰²	97.1		%	96.4-98.8
Hgb A2 ⁰²	2.9		%	1.8-3.2
Hgb S ⁰²	0.0		%	0.0

Interpretation: ⁰²	Normal hemoglobin present; no hemoglobin variant or beta thalassemia identified. Note: Alpha thalassemia may not be detected by the Hgb Fractionation Cascade panel. If alpha thalassemia is suspected, Labcorp offers Alpha-Thalassemia DNA Analysis (#511172).			
ABO Grouping ⁰¹	A			
Rh Factor ⁰¹	Positive			
	Please note: Prior records for this patient's ABO / Rh type are not available for additional verification.			

Date Collected: 03/20/2024

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

CBC, Platelet Ct, and Diff ⁰¹			
WBC ⁰¹	6.5	x10E3/uL	3.4-10.8
RBC ⁰¹	5.46	x10E6/uL	4.14-5.80
Hemoglobin ⁰¹	16.4	g/dL	13.0-17.7
Hematocrit ⁰¹	48.1	%	37.5-51.0
MCV ⁰¹	88	fL	79-97
MCH ⁰¹	30.0	pg	26.6-33.0
MCHC ⁰¹	34.1	g/dL	31.5-35.7
RDW ⁰¹	12.2	%	11.6-15.4
Platelets ⁰¹	212	x10E3/uL	150-450
Neutrophils ⁰¹	62	%	Not Estab.
Lymphs ⁰¹	29	%	Not Estab.
Monocytes ⁰¹	7	%	Not Estab.
Eos ⁰¹	2	%	Not Estab.
Basos ⁰¹	0	%	Not Estab.
Neutrophils (Absolute) ⁰¹	4.0	x10E3/uL	1.4-7.0
Lymphs (Absolute) ⁰¹	1.9	x10E3/uL	0.7-3.1
Monocytes(Absolute) ⁰¹	0.5	x10E3/uL	0.1-0.9
Eos (Absolute) ⁰¹	0.1	x10E3/uL	0.0-0.4
Baso (Absolute) ⁰¹	0.0	x10E3/uL	0.0-0.2
Immature Granulocytes ⁰¹	0	%	Not Estab.
Immature Grans (Abs) ⁰¹	0.0	x10E3/uL	0.0-0.1
Urinalysis Gross Exam ⁰¹			
Specific Gravity ⁰¹	1.007		1.005-1.030
pH ⁰¹	6.5		5.0-7.5
Urine-Color ⁰¹	Yellow		Yellow
Appearance ⁰¹	Clear		Clear
WBC Esterase ⁰¹	Negative		Negative
Protein ⁰¹	Negative		Negative/Trace
Glucose ⁰¹	Negative		Negative
Ketones ⁰¹	Negative		Negative
Occult Blood ⁰¹	Negative		Negative
Bilirubin ⁰¹	Negative		Negative
Urobilinogen,Semi-Qn ⁰¹	0.2	mg/dL	0.2-1.0
Nitrite, Urine ⁰¹	Negative		Negative
Microscopic Examination ⁰¹	Microscopic not indicated and not performed.		

Disclaimer
The Previous Result is listed for the most recent test performed by Labcorp in the past 5 years where there is sufficient patient demographic data to match the result to the patient. Results from certain tests are excluded from the Previous Result display.

Icon Legend
▲ Out of Reference Range ■ Critical or Alert

Performing Labs
01: SE - Labcorp Seattle, 550 17th Avenue Ste 300, Seattle, WA 98122-5789 Dir: Daniel Toweill, MD
02: BN - Labcorp Burlington, 1447 York Court, Burlington, NC 27215-3361 Dir: Sanjai Nagendra, MD
For Inquiries, the physician may contact Branch: 206-861-7000 Lab: 206-861-7000

Patient Details	Physician Details	Specimen Details
13144, Donor	J KUAN	Specimen ID: 080-129-3460-0
4915 25TH AVE NE STE 204, SEATTLE, WA, 981055668	Seattle Sperm Bank	Control ID: L2402204989
	4915 25th Ave Ne Ste 204, SEATTLE, WA, 98105	Alternate Control Number: L2402204989
Phone:	Phone: 206-588-1484	Date Collected: 03/20/2024 1318 Local
Date of Birth: [REDACTED]	Account Number: 46857540	Date Received: 03/21/2024 0000 ET
Age: [REDACTED]	Physician ID:	Date Entered: 03/21/2024 0434 ET
Sex: Male	NPI: 1891885745	Date Reported: 03/27/2024 1008 ET
Patient ID:		
Alternate Patient ID:		