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CLIA: 22D0957540



Patient Information:
10942, Donor

Sex: M
MR#:
Patient#: FT-PT8859830

Accession:
FT-7213903
Test#: FT-TS14966961
Specimen Type: Blood (EDTA)
Collected: Oct 01, 2024

Partner Information:
Not Tested

Accession:
N/A

Physician:
Kuan, James
ATTN: Kirk, Ashley
Phoenix Sperm Bank
4915 25th Avenue NE, Ste 204W
Seattle, WA 98105
Phone: (206) 588-1484

Laboratory:
Fulgent Therapeutics LLC
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Lawrence M. Weiss, MD
Report Date: **Oct 15, 2024**

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	10942, Donor	Partner
Joubert syndrome 9 <i>CC2D2A</i>	AR	⊕ Carrier c.3774dup (p.Glu1259*)	N/A
Usher syndrome, type 1D <i>CDH23</i>	AR	⊕ Carrier c.6050-9G>A (p.?)	N/A

INTERPRETATION:

Notes and Recommendations:

- Possible digenic inheritance has been reported for Usher syndrome type 1D/F with variants in both CDH23 and PCDH15 genes (PubMed: [15537665](#), [35353227](#), [30531642](#); OMIM: [605516](#), [605514](#)). Correlation with clinical and family history is recommended.
- Based on these results, this individual is positive for carrier mutations in 2 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: 10942, Donor; Sex: M;
DOB: [REDACTED]; MR#:

Accession#: FT-7213903; FD Patient#: FT-PT8859830;
DocID: FT-TS14966961AA; PAGE 1 of 6



JOUBERT SYNDROME 9

Patient	10942, Donor	Partner
Result	 Carrier	N/A
Variant Details	CC2D2A (NM_001080522.2) c.3774dup (p.Glu1259*)	N/A

What is Joubert syndrome 9?

Joubert syndrome is a group of related disorders characterized by the underdevelopment of the cerebella vermis of the brain and malformed brain stem, low muscle tone, developmental delay and intellectual disabilities. Distinctive facial features, including a broad forehead, arched eyebrows, droopy eyelids, widely spaced eyes, low-set ears, and a triangular-shaped mouth, are also characteristic. Other symptoms may include, retinal dystrophy, kidney and liver disease, extra fingers and toes, and endocrine issues.

What is my risk of having an affected child?

Joubert syndrome 9 is inherited in an autosomal recessive manner. This means that if both parents are carriers, their risk of having an affected child is 1 in 4 (25%).

What kind of medical management is available?

The treatment for Joubert syndrome is symptomatic and supportive. Developmental delays are usually treated with physical therapy, occupational therapy, speech therapy, and infant stimulation. Annual screening is recommended for liver kidney and retinal abnormalities.

What mutation was detected?

The detected heterozygous variant was NM_001080522.2:c.3774dup (p.Glu1259*). This frameshift variant, p.Glu1259*fs (also reported as p.E1259fsX1 or p.Glu1295*), 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 CC2D2A-related conditions (PubMed: [19777577](#), [30919572](#), [18950740](#), [26729329](#)). This variant has been reported in compound heterozygous state in two individuals with Joubert syndrome and two with a clinical diagnosis of Meckel-Gruber syndrome (PubMed: [21068128](#), [21866095](#), [24706459](#), [26092869](#)). In addition, truncating variants downstream of this position have been reported to be pathogenic (PubMed: [22241855](#), [19466712](#), [26092869](#)). This variant is classified as "Pathogenic" in ClinVar, with multiple submitters in agreement (Variation ID: 56309). The laboratory classifies this variant as pathogenic.



USHER SYNDROME, TYPE 1D

Patient	10942, Donor	Partner
Result	 Carrier	N/A
Variant Details	CDH23 (NM_022124.6) c.6050-9G>A (p.?)	N/A

What is Usher syndrome, type 1D?

Usher syndrome is a group of conditions characterized by partial or total hearing loss and vision loss that worsens over time. The different types are distinguished by the severity of hearing loss, balance issues, and age of onset; which ranges from congenital hearing loss with vision loss beginning in childhood or adolescence to normal hearing and vision until late childhood or adolescence that gradually begins to dissipate. Some children also experience vestibular dysfunction that affects balance and may impact motor development.

What is my risk of having an affected child?

Usher syndrome, type 1D 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?

Currently, there is no cure for Usher syndrome. The best treatment involves early identification so that educational programs can begin as soon as possible. Typically, treatment will include hearing aids, assistive listening devices, cochlear implants, or other communication methods such as American Sign Language, orientation and mobility training, communication services, and independent-living training that may include Braille instruction, low-vision services, or auditory training.

What mutation was detected?

The detected heterozygous variant was NM_022124.6:c.6050-9G>A (p.?). This intronic variant, c.6050-9G>A, is predicted by multiple splice site models (3 of 5), to result in the creation of a new acceptor site. This variant has been previously reported in patients with Usher syndrome (PubMed: [11857743](#), [25404053](#), [24498627](#), [32467589](#), [35020051](#), [31546658](#)). Exon trapping experiments have demonstrated this variant creates a new acceptor site and results in an insertion of 7-nucleotide which causes a frameshift (PubMed: [11857743](#)). This variant has been reported to segregate with disease in at least one family with Usher Syndrome (PubMed: [31546658](#)). Functional analysis of this variant indicates that this change results in exon skipping and generated aberrantly spliced transcripts (PubMed: [31546658](#)). This variant is classified as "Pathogenic" or "Likely Pathogenic" in ClinVar, with multiple submitters in agreement (Variation ID: 46001). The laboratory classifies this variant as 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, PUVK, 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, RARSN, RARS2, RDH12, RLB1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RPE65, RPGRIP1L, RTEL1, RXYLT1, RYR1, SACS, SAMD9, SAMHD1, SCQ2, 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, 99.57% and 99.52% 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.

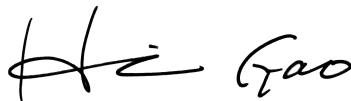
Gene Specific Notes and Limitations

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. 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. CFTR variants primarily associated with CFTR-related isolated congenital bilateral absence of the vas deferens and CFTR-related pancreatitis are not included in this analysis. CFTR variants with insufficient evidence of being cystic fibrosis mutations will not be reported either. **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 the clinical findings, biochemical profile, and family history of each patient. LR-PCR is not routinely ordered for NM_000500.9:c.955C>T (p.Gln319Ter). Individuals with c.955C>T (p.Gln319Ter) will be reported as a Possible Carrier indicating that the precise nature of the variant has not been determined by LR-PCR and that the variant may occur in the CYP21A2 wild-type gene or in the CYP21A1P pseudogene. The confirmation test is recommended if the second reproductive partner is tested positive for variants associated with classic CAH. **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. NPHS2: If detected, the variant NM_014625.3:c.686G>A (p.Arg229Gln) will not be reported as this variant is not significantly associated with disease when homozygous or in the compound heterozygous state with variants in exons 1-6 of NPHS2. 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 in exon 7 and copy number variations in exons 7-8 of the SMN1 gene (NM_022874.2). Sequencing and deletion/duplication analysis are not performed on any other region in this gene. About 5%-8% of the population have two copies of SMN1 on a single chromosome and a deletion on the other chromosome, known as a [2+0] configuration (PubMed: [20301526](#)). The current testing method cannot directly detect carriers with a [2+0] SMN1 configuration but can detect linkage between the silent carrier allele and certain population-specific single nucleotide changes. As a result, a negative result for carrier testing greatly reduces but does not eliminate the chance that a person is a carrier. Only abnormal results will be reported. 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). VPS45: LoF is not a known disease mechanism. 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:



Dr. Harry Gao, DABMG, FACMG on 10/15/2024
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:

10942, Donor

DOB: [REDACTED]

Sex: M

MR#:

Patient#: FT-PT8859830

Partner Information:

Not Tested

Physician:

Kuan, James

ATTN: Kirk, Ashley

Phoenix Sperm Bank

4915 25th Avenue Northeast

Ste 204

Seattle, 98105

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Laboratory:

Fulgent Therapeutics LLC

CAP#: 8042697

CLIA#: 05D2043189

Laboratory Director:

Dr. Amar Jariwala

Report Date: **Dec 15, 2025**

Accession:

FT-7213903

Test#: FT-TS15424852

Specimen Type: Blood (EDTA)

Collected: Oct 01, 2024

Accession:

N/A

FINAL RESULTS



No carrier mutations identified

TEST PERFORMED

Single Gene Carrier Screening: GNRHR

(1 Gene Panel: *GNRHR*; 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 - Gene

This analysis was run using the Custom Beacon Preconception Carrier Screening Panel gene list. 1 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.

GNRHR

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 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.

15 Crawford St., STE 100
Needham, MA 02494
(p) 626-350-0537
(f) 626-454-1667
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 'Yan Meng'.

Yan Meng, Ph.D., CGMB, FACMG on 12/15/2025
Laboratory Director, Fulgent

DISCLAIMER:

This test was developed, performed, and its performance characteristics determined by **Fulgent Therapeutics LLC** (CAP# 8042697, CLIA# 05D2043189), 4399 Santa Anita Ave., El Monte, CA 91731. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at **626-350-0537** or 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: 10942, Donor; Sex: M;
DOB: [REDACTED] MR#:

Accession#: FT-7213903; FD Patient#: FT-PT8859830;
DocID: FT-TS15424852AA; **PAGE 3 of 3**

Report Status FINAL
Route 2017 Ordered by:
Phoenix Sperm Bank
1492 S Mill Ave
Suite 306
Tempe, AZ 85281

James Kuan, MD

Patient Information:

10942, DONOR

Account: 18131
ID/MR#: 10942
Patient Lab ID:
66rd9d0dc610ad2bb0409b31

Collected: 10/01/2024 11:53 AM
Received: 10/02/2024 11:45 AM
Reported: 10/11/2024 05:05 PM

Order
42
47

Patient Phone: 602-888-7255

PL

GENETICS

Accession #:
CG240010605
Cell Type/Source:
Blood

Clinician Provided Information:
DONOR TESTING

Chromosome Analysis: Routine Blood

Analysis Details:

Metaphases/Cells Counted : 20
Metaphases/Cells Analyzed : 5
Metaphases Karyotyped : 3

PV

Results:

NORMAL MALE KARYOTYPE

PV

46,XY

PV

Interpretation:

Normal

PV

Normal karyotype at the band level 550 or above as determined by the trypsin-Giemsa method. There was no evidence for a chromosome abnormality within the limits of the band level and technology utilized in this study.

PHA-stimulated lymphocyte chromosome analysis is an accurate technique to detect many constitutional chromosome abnormalities. More extensive investigation may be required to detect mosaicism or subtle structural rearrangement. It also should be noted that this type of testing does not rule out the possibility of mendelian, mitochondrial, multifactorial or environmental etiologies.

Comments:

remote: iic,lds

PV

Cytogenetics Director:

PV



Electronically signed by Liwen Lai R PhD, DABMGG, FACMG R
Verified 10/11/24

Report Status FINAL

Route 2017 Ordered by:
Phoenix Sperm Bank
1492 S Mill Ave
Suite 306
Tempe, AZ 85281



**Sonora Quest
Laboratories™**

A Subsidiary of Laboratory Sciences of Arizona

James Kuan, MD

Patient Information:

10942, DONOR

Account: 18131
ID/MR#: 10942
Patient Lab ID:
66rd9d0dc610ad2bb0409b31

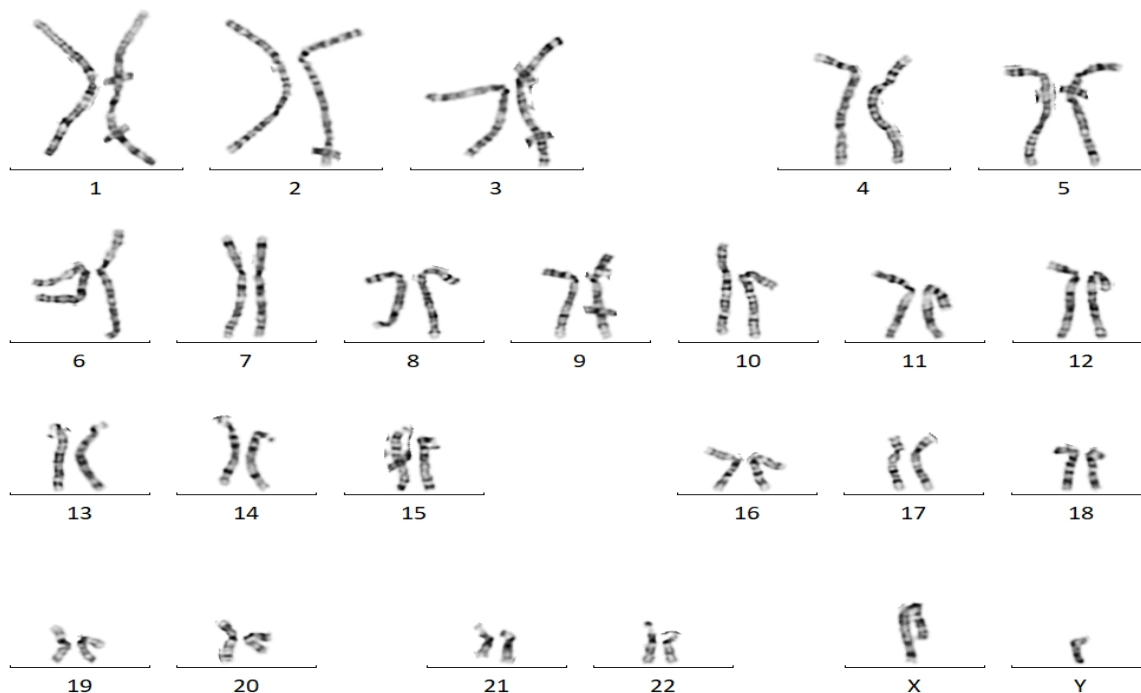
Collected: 10/01/2024 11:53 AM
Received: 10/02/2024 11:45 AM
Reported: 10/11/2024 05:05 PM

Order

42

47

Patient Phone: 602-888-7255



Tests Ordered: Chromosome Analysis: Routine Blood

Unless otherwise noted, testing performed by: Sonora Quest Laboratories, 424 S 56th St, Phoenix, AZ 85034 800.766.6721
Testing noted as PV performed by: Genetics/Genomics Div., Sonora Quest Laboratories, 424 S. 56th St, Phoenix, AZ 85034 602.685.5700

End of Report

10942, DONOR Order #: 181310000142 / NL105334447 - FINAL Report

L=Low, H=High, C=Critical Abnormal, CL=Critical Low, CH=Critical High, *=Comment

Distribution #: 746455258-40890974



Result Report

Produced by AutoDist On 10/11/2024 05:09 PM

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Date Collected: 10/01/2024

Date Received: 10/02/2024

Date Reported: 10/05/2024

Fasting: Not Given

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

Date Collected: 10/01/2024

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

Test	Current Result and Flag		Previous Result and Date	Units	Reference Interval
Glucose ⁰¹	80			mg/dL	70-99
Uric Acid ⁰¹	4.9			mg/dL	3.8-8.4
Therapeutic target for gout patients: <6.0					
▲ BUN ⁰¹	23	High		mg/dL	6-20
Creatinine ⁰¹	1.14			mg/dL	0.76-1.27
eGFR	90			mL/min/1.73	>59
Calcium ⁰¹	9.6			mg/dL	8.7-10.2
Protein, Total ⁰¹	6.9			g/dL	6.0-8.5
Albumin ⁰¹	4.7			g/dL	4.3-5.2
Bilirubin, Total ⁰¹	1.2			mg/dL	0.0-1.2
Alkaline Phosphatase ⁰¹	62			IU/L	44-121
LDH ⁰¹	161			IU/L	121-224
AST (SGOT) ⁰¹	20			IU/L	0-40
ALT (SGPT) ⁰¹	12			IU/L	0-44
Cholesterol, Total ⁰¹	144			mg/dL	100-199
Triglycerides ⁰¹	114			mg/dL	0-149
HDL Cholesterol ⁰¹	47			mg/dL	>39
LDL Chol Calc (NIH)	76			mg/dL	0-99
LDL/HDL Ratio	1.6			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	97.5			%	96.4-98.8
Hgb A2 ⁰²	2.5			%	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	O				
Rh Factor ⁰¹	Negative				
Please note: Prior records for this patient's ABO / Rh type are not available for additional verification.					

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

CBC, Platelet Ct, and Diff ⁰¹			
WBC ⁰¹	5.5	x10E3/uL	3.4-10.8
RBC ⁰¹	5.07	x10E6/uL	4.14-5.80
Hemoglobin ⁰¹	15.0	g/dL	13.0-17.7
Hematocrit ⁰¹	46.3	%	37.5-51.0
MCV ⁰¹	91	fL	79-97
MCH ⁰¹	29.6	pg	26.6-33.0
MCHC ⁰¹	32.4	g/dL	31.5-35.7
RDW ⁰¹	12.1	%	11.6-15.4
Platelets ⁰¹	269	x10E3/uL	150-450
Neutrophils ⁰¹	61	%	Not Estab.
Lymphs ⁰¹	29	%	Not Estab.
Monocytes ⁰¹	7	%	Not Estab.
Eos ⁰¹	2	%	Not Estab.
Basos ⁰¹	1	%	Not Estab.
Neutrophils (Absolute) ⁰¹	3.4	x10E3/uL	1.4-7.0
Lymphs (Absolute) ⁰¹	1.6	x10E3/uL	0.7-3.1
Monocytes(Absolute) ⁰¹	0.4	x10E3/uL	0.1-0.9
Eos (Absolute) ⁰¹	0.1	x10E3/uL	0.0-0.4
Baso (Absolute) ⁰¹	0.1	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	1.026		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	Trace		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: PDLCA - Labcorp Phoenix, 5005 S 40th Street Ste 1200, Phoenix, AZ 85040-2969 Dir: Earle Collum, MD
02: BN - Labcorp Burlington, 1447 York Court, Burlington, NC 27215-3361 Dir: Sanjai Nagendra, MD
For Inquiries, the physician may contact Branch: 888-522-2677 Lab: 800-788-9743

Patient Details	Physician Details	Specimen Details
10942, Donor	J KUAN	Specimen ID: 275-705-5505-0
	Phoenix Sperm Bank	Control ID: L2407402826
	1492 S Mill Ave Suite 306, Tempe, AZ, 85281	Alternate Control Number: L2407402826
Phone:		Date Collected: 10/01/2024 1155 Local
Date of Birth: [REDACTED]	Phone: 602-888-7255	Date Received: 10/02/2024 0000 ET
Age: [REDACTED]	Account Number: 02003540	Date Entered: 10/02/2024 0549 ET
Sex: Male	Physician ID:	Date Reported: 10/05/2024 0908 ET
Patient ID:	NPI: 1891885745	
Alternate Patient ID:		