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fulgent

Patient Information:

DOB:
Sex: M
MR#: BFA0223

Partner Information:
Not Tested

Physician:
Shaikly, Valerie
ATTN: Shaikly, Valerie
Fertility Genetics
1 Lanswood Park
Elmstead Market, Essex CO7 7FD GB
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Laboratory:
Fulgent Therapeutics LLC
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Amar Jariwala
Report Date: **Sep 09,2026**

Accession:

Accession:
N/A

Specimen Type: Saliva Swab
Collected: Aug 18,2026

FINAL RESULTS

Carrier for genetic conditions in **multiple** genes.

TEST PERFORMED

176 Matched Fors Male with XL
(177 Gene Panel; gene sequencing with deletion and duplication analysis)

Condition and Gene	Inheritance		Partner
Krabbe disease *GALC*	AR	Carrier Deletion of Exons 11-17	N/A
Mucolipidosis III gamma *GNPTG*	AR	Carrier Deletion of Exons 4-part of exon 6	N/A

INTERPRETATION:

Notes and Recommendations:

- 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 a 3 nucleotide (CGG) repeat sequence in the FMR1 gene was performed to screen for the carrier status for Fragile X Syndrome. The repeat size detected was: 20 repeats. These results are within the normal range. Therefore, this individual is not considered a carrier for Fragile X Syndrome.
- 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.
- Gene specific notes and limitations may be present. See below.
- Genetic counseling is recommended. Contact your physician about the available options for genetic counseling.

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KRABBE DISEASE

Patient		Partner
Result	Carrier	N/A
Variant Details	***GALC*** (NM_000153.4) Deletion of Exons 11-17	N/A

What is Krabbe disease?

Krabbe disease (globoid cell leukodystrophy) is a degenerative disorder caused by a deficiency of the enzyme galactosylceramidase. This deficiency leads to the demyelination of nerve cells. Most individuals have infantile-onset Krabbe disease, presenting before 12 months of age with irritability, spasticity, developmental delay, feeding difficulties, vision loss, and seizures. Individuals with a later-onset form of Krabbe disease can be clinically normal until weakness, vision loss, and intellectual regression become apparent. The onset and progression of the later-onset forms can be quite variable, even among siblings.

What is my risk of having an affected child?

Krabbe disease 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 of individuals with infantile-onset Krabbe disease who are diagnosed in a late stage is limited to supportive care to control irritability and spasticity. Hematopoietic stem cell transplantation (HSCT) may improve and preserve cognitive function in presymptomatic infants and older individuals with mild symptoms; however, many show progressive deterioration of the peripheral nervous system.

What mutation was detected?

This exonic deletion removes the last 7 exons. It is predicted to result in the loss of function of the protein product. There's sufficient evidence that loss of function in this gene is a known disease mechanism for Krabbe (PubMed: 16607461, 20886637, 17579360, 27780934). This variant has been identified in the homozygous and compound heterozygous state in several unrelated individuals affected with Krabbe disease and has been shown to segregate with disease in multiple families (PubMed: 26795590, 36057781, 23197103, 38837642, 35286032, 36781956). The allele frequency of this deletion in the general population is 0.037%. The scope of the performed analysis is limited to the ordered genes and is not designed to determine the exact breakpoints or boundaries of copy number variants. This variant may or may not represent part of a larger event involving other potentially clinically relevant genes not assessed by this test. The laboratory classifies this variant as pathogenic.

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MUCOLIPIDOSIS III GAMMA

Patient		Partner
Result	Carrier	N/A
Variant Details	***GNPTG*** (NM_032520.5) Deletion of Exons 4-part of exon 6	N/A

What is Mucolipidosis III gamma?

Mucolipidosis III gamma is a progressive disorder characterized by joint stiffening and multiple skeletal abnormalities. Individuals with mucolipidosis III gamma typically are slow growing during childhood and of short stature as adults. Symptoms commonly start manifesting at the age of three. A characteristic symptom is stiffness of the fingers, which eventually leads to a claw-like appearance of the hands. Other characteristics are inward-bend knees, gradual thickening and coarsening of the skin, deformations in shoulders and hips as well as heart defects, which may increase the risk of sudden cardiac arrest. Other complications may include breathing problems and developmental delay, particularly with regard to motor skills. Intellect is typically not affected.

What is my risk of having an affected child?

Mucolipidosis III gamma 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?

Progression of mucolipidosis III gamma cannot be stopped and symptoms may not be reversed. Affected individuals may need supportive physiotherapy to help with the symptoms. Children often need assistance in school due to physical limitations. In later stages progressing stiffening results typically in frequent pain and may need medical treatment and in some cases hip replacement may be necessary and helpful. In cases with severe heart defects, surgery may be needed.

What mutation was detected?

This exonic deletion is predicted to lead to an out-of-frame shift transcript and introduce a premature stop codon at least 50 nucleotides upstream of the canonical donor splice site of the penultimate exon, resulting 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 mucolipidosis III gamma (PubMed: 29170090, 29872134, 35005816).This variant has not been reported as associated with a clinical condition in the Human Gene Mutation Database (HGMD).This copy number variant is absent from controls in the gnomAD structural variants database. The Broad Institute gnomAD structural variants database (SVs) was used for this analysis. The laboratory classifies this variant as likely pathogenic.

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GENES TESTED:

176 Matched Fors Male with XL - 177 Genes

This analysis was run using the 176 Matched Fors Male with XL gene list. 177 genes were tested with 99.2% of targets sequenced at >20x coverage. For more gene-specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

ABCC8, ABCD1, ACADM, ACADS, ACADVL, ADA, AGA, AGL, AGXT, AIRE, ALDH3A2, ALDOB, ALG6, ALMS1, ALPL, AMT, ARG1, ARSA, ASL, ASPA, ASS1, ATM, ATP7A, ATP7B, BBS1, BBS10, BBS12, BBS2, BCKDHA, BCKDHB, BCS1L, BLM, BTD, CAPN3, CBS, CFTR, CLN3, CLN5, CLN6, CLN8, CLRN1, COL4A3, COL4A4, CPS1, CPT1A, CPT2, CRYL1, CTNS, CTSK, CYP11B1, CYP21A2, CYP27A1, DBT, DHCR7, DLD, DMD, DYSF, ELP1, ERCC6, ERCC8, EVC, EVC2, FAH, FANCA, FANCC, FKRP, FKTN, FMR1, G6PC, GAA, GALC, GALK1, GALT, GBA, GCDH, GJB2, GJB6, GLA, GLB1, GLDC, GNE, GNPTAB, GNPTG, GRHPR, HADHA, HBA1, HBA2, HBB, HEXA, HEXB, HGSNAT, HLCS, HMGCL, HOGA1, HSD17B4, HYLS1, IDS, IDUA, IL2RG, IVD, KCNJ11, LAMA2, LAMA3, LAMB3, LAMC2, LIPA, LRPPRC, MAN2B1, MCOLN1, MEFV, MESP2, MKS1, MLC1, MMAA, MMAB, MMACHC, MPI, MUT, MYO7A, NAGLU, NBN, NEB, NPC1, NPC2, NPHS1, NPHS2, NR0B1, OPA3, OTC, PAH, PC, PCCA, PCCB, PCDH15, PEX1, PEX10, PEX12, PEX2, PEX6, PEX7, PKHD1, PMM2, POMGNT1, PPT1, PROP1, PTS, RMRP, RS1, RTEL1, SACS, SGCA, SGCB, SGCD, SGCG, SGSH, SLC12A6, SLC17A5, SLC22A5, SLC26A2, SLC26A4, SLC37A4, SMN1, SMPD1, STAR, TAT, TCIRG1, TGM1, TH, TMEM216, TPP1, TTPA, USH1C, USH2A, VPS13B, XPA, XPC, ZFYVE26

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, 99.34% and 99.21% 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

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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.

Gene Specific Notes and Limitations

BTD: If detected, the variant NM_001370658.1:c.1270G>C (p.Asp424His) will not be reported as this variant is associated with low disease penetrance and is primarily associated with reduced enzyme activity when homozygous. *CFTR:* Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) variant 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 causing cystic fibrosis 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, 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. *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. CYP21A2 variants primarily associated with non-classic congenital adrenal hyperplasia (CAH) are not included in this analysis (PubMed: 23359698). The variants associated with non-classic disease, including but not limited to c.188A>T (p.His63Leu), c.844G>T (p.Val282Leu), c.1174G>A (p.Ala392Thr), and c.1360C>T (p.Pro454Ser) will not be reported. 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. *DMD:* Single exon deletion/duplication analysis is limited to exons with >1 patient reported in the UMD database (http://www.umd.be/DMD/W_DMD/index.html), accessed Dec 29,2020 and all out-of-frame exons after exon 3. This includes deletion of exon 1, and duplication of exon 2, and del/dup for exons 3,6~8,11,12,17~22,43~46,48,50~56,58~63,65~70,75,76 and 78. Single-exon detection is limited to blood samples. *FMR1:* The exact size of alleles >200 CGG repeats cannot be determined; these alleles are pathogenic for X-Linked Fragile X Syndrome. Alleles with <10 repeats may fail to amplify; these alleles are benign. The repeat length for this gene may vary by +/- 1 repeat unit. Methylation is not analyzed. RP-PCR analysis of the FMR1 promoter is not routinely performed in males. Small degrees of size mosaicism, including gonadal mosaicism, may not be detected. *GALT:* The D2 "Duarte" allele is not included in this analysis. While this allele can cause positive newborn screening results, it is not known to cause clinical symptoms in any state (PubMed: 25473725, 30593450). *GBA:* The current testing method may not be able to reliably detect certain pathogenic variants in the GBA gene due to homologous recombination between the pseudogene and the functional gene. *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. *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. *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.

SIGNATURE:

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Zhenbin Chen, Ph.D., CGMB, FACMG on **09/09/2026**
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.

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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