



Patient Information:

20053, Donor
DOB: [REDACTED]
Sex: M
MR#: [REDACTED]
Patient#: FT-PT8963863

Partner Information:

Not Tested

Physician:

Kuan, JAMES
ATTN: Kuan, JAMES
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Laboratory:

Fulgent Therapeutics LLC
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Amar Jariwala
Report Date: **Feb 11, 2025**

Accession:

FT-7138282
Test#: FT-TS15072020
Specimen Type: Blood (EDTA)
Collected: Jan 22, 2025

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	20053, Donor	Partner
Wilson disease <i>ATP7B</i>	AR	⊕ Carrier c.2128G>A (p.Gly710Ser)	N/A
CEP152-related disorders <i>CEP152</i>	AR	⊕ Carrier c.2034T>G (p.Tyr678*)	N/A
WNT10A-related ectodermal dysplasias <i>WNT10A</i>	AR	⊕ Carrier c.682T>A (p.Phe228Ile)	N/A

INTERPRETATION:

Notes and Recommendations:

- **PLEASE NOTE: Heterozygous carriers of a WNT10A variant may be at risk of autosomal dominant selective tooth agenesis-4 (STHAG4) (PubMed: 29772684; OMIM: 606268). Autosomal dominant phenotypes have also been described to include dry skin, abnormal sweating, nail abnormalities and sparse hair in some patients (PubMed: 20301291, 34184264; OMIM: 150400). As such, correlation with clinical and family history is recommended. Consultation with a medical geneticist and/or other specialist is recommended.**
- 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>)

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Patient: 20053, Donor; Sex: M;
DOB: [REDACTED] MR#:

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WILSON DISEASE

Patient	20053, Donor	Partner
Result	 Carrier	N/A
Variant Details	ATP7B (NM_000053.4) c.2128G>A (p.Gly710Ser)	N/A

What is Wilson disease?

Wilson disease is a disorder that affects the liver's ability to remove excess copper in the body. Normally, a healthy amount of copper is absorbed by the body through diet. When the liver is impaired and cannot properly excrete copper, the buildup over time can lead to symptoms such as liver disease, psychiatric disturbance, and neurologic disorder. Ages of onset vary, with symptoms most commonly reported between the ages of 5 to 35.

What is my risk of having an affected child?

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

What kind of medical management is available?

Medical management is specific to each individual and should be discussed with your doctor. To treat Wilson disease, lifelong management is required. Administration of chelating agents (medications) to remove excess copper and reducing dietary intake of copper may help to prevent disease progression. Liver transplantation may also be required in severe cases of liver damage. Biannual surveillance is strongly recommended to assess the progression of the disease and/or treatment. Routine monitoring should include blood tests, physical examinations, and liver function tests.

What mutation was detected?

The detected heterozygous variant was NM_000053.4:c.2128G>A (p.Gly710Ser). This variant, p.Gly710Ser, was previously identified in the homozygous and compound heterozygous states in the ATP7B gene of multiple patients with Wilson disease (PubMed: [8938442](#), [10406672](#), [27398169](#), [11857545](#), [31169307](#), [31708252](#), [34400371](#)). Other missense changes in the same codon as this variant (p.Gly710Ala, p.Gly710Val, p.Gly710Arg, p.Gly710Cys) have been identified in patients with Wilson disease (PubMed: [16088907](#), [24253677](#), [9887381](#), [27982432](#), [8931691](#)). In vitro functional studies demonstrated that this variant leads to significantly decreased copper uptake by the ATP7B transporter (PubMed: [22240481](#)). The laboratory classifies this variant as pathogenic.



CEP152-RELATED DISORDERS

Patient	20053, Donor	Partner
Result	+ Carrier	N/A
Variant Details	CEP152 (NM_014985.4) c.2034T>G (p.Tyr678*)	N/A

What is CEP152-related disorders?

The CEP152 gene is associated with two conditions: Autosomal recessive primary microcephaly 9 and Seckel syndrome 5.

- Autosomal recessive primary microcephaly 9 is an inherited condition in which infants are born with a very small head and brain. An affected individual's head and brain will continue to grow throughout their life, but it will remain much smaller than normal. Most individuals will have mild to moderate intellectual disability, delayed language acquisition, and delayed motor skills. Some may have mild seizures, attention and behavior concerns, and short stature. Primary microcephaly typically does not affect any other major organ systems.
- Seckel syndrome 5 is a primordial dwarfism disorder of prenatal onset characterized by intrauterine growth restriction, dwarfism, and severe microcephaly with moderate to severe intellectual disability. Individuals with this condition also have a characteristic "bird-headed" facial appearance which features a sloping forehead, beak-like nose, large eyes, narrow face, differently shaped ears, and/or small jaw. Clinodactyly, hip dysplasia, and radial dislocation, and other skeletal abnormalities are common findings. Males with Seckel syndrome may have cryptorchidism and affected females may have clitoromegaly. In addition, affected children may have increased body hair. In some cases, hematological problems may occur including anemia, pancytopenia, and acute myeloid leukemia.

What is my risk of having an affected child?

CEP152-related disorders are 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?

Each condition is managed differently based on its symptoms.

- There are no specific treatments for microcephaly. Early intervention that includes speech, physical, and occupational therapies can support cognitive and motor development. Medications, such as anticonvulsants, may stabilize seizures. The prognosis depends on the severity and manifestations of the condition but is generally good.
- For Seckel syndrome, affected individuals should be monitored, assessed, and appropriately treated to manage the symptoms of the condition. Treatment and management is directed toward the specific symptoms present in each individual, including intellectual disability, skeletal abnormalities, genital differences, and hematological problems.

What mutation was detected?

The detected heterozygous variant was NM_014985.4:c.2034T>G (p.Tyr678*). 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 is sufficient evidence that loss of function in this gene is a known disease mechanism for primary microcephaly and Seckel syndrome (PubMed: [34402213](#), [20598275](#), [21131973](#)). This variant has been reported in the compound heterozygous state in an individual with Seckel syndrome (PubMed: [21131973](#)). This variant is classified as "Pathogenic" or "Likely Pathogenic" in ClinVar, with multiple submitters in agreement (Variation ID: 158240). The laboratory classifies this variant as pathogenic.



WNT10A-RELATED ECTODERMAL DYSPLASIAS

Patient	20053, Donor	Partner
Result	 Carrier	N/A
Variant Details	WNT10A (NM_025216.3) c.682T>A (p.Phe228Ile)	N/A

What is WNT10A-related ectodermal dysplasias?

The WNT10A gene produces proteins that aid in the formation of several types of tissues that arise from an embryonic cell layer called the ectoderm. There are several conditions associated with this gene including Schopf-Schulz-Passarge syndrome and Odonto-onycho-dermal dysplasia.

- Schopf-Schulz-Passarge syndrome (SSPS) and Odonto-onycho-dermal dysplasia (OODD) are rare ectodermal dysplasias characterized by hypodontia, keratoderma, nail dystrophy, and hair abnormalities. Individuals with SPSS also have multiple eyelid cysts, and hypotrichosis. Dry hair, smooth tongue with marked reduction of fungiform and filiform papillae, hyperhidrosis of palms and soles, and hyperkeratosis of the skin are additional features seen in individuals with OODD. For both conditions, some features may not be present until adulthood, causing diagnoses to be delayed in some cases.

What is my risk of having an affected child?

Schopf-Schulz-Passarge syndrome and Odontoonychodermal dysplasia are 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?

Each condition is managed differently based on its symptoms. The treatment of SSPS and OODD is symptomatic. Standard dental care is indicated in most cases. Eyelid cysts seen in those with SSPS may be improved by electrocautery. If necessary, counseling and psychological support and dermatological examination to detect non-melanoma skin cancer should be offered.

What mutation was detected?

The detected heterozygous variant was NM_025216.3:c.682T>A (p.Phe228Ile). This variant, p.Phe228Ile, has been reported in many individuals with isolated oligodontia (PubMed: [22581971](#), [24449199](#), [24311251](#)). This variant has been reported in affected individuals in the homozygous, compound heterozygous, and heterozygous state, but also has been identified in unaffected individuals (PubMed: [36294409](#), [32618450](#), [34593752](#)). This variant is prevalent and is known to be associated with reduced penetrance (PubMed: [28976000](#), [24700731](#)), suggesting all carriers may not show symptoms of the disease. This variant is classified as "Pathogenic" or "Likely Pathogenic" by multiple clinical laboratories in ClinVar (Variation ID: 4462). 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, GRHRP, 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, MKK5, 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, 99.61% and 99.49% 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, 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). **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, CGMB on 2/11/2025
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 tested on any Beacon panel, please visit the following link:

[Beacon Expanded Carrier Screening Supplemental Table](#)





Patient Information	Specimen Information	Client Information
20053, DONOR DOB: [REDACTED] AGE: [REDACTED] Gender: M Fasting: U Phone: 725.257.0900 Patient ID: 20053	Specimen: LV393354Y Requisition: 0000040 Collected: 01/22/2025 / 14:11 PST Received: 01/23/2025 / 02:00 PST Reported: 01/30/2025 / 08:35 PST	Client #: 88807473 MAIL500 FOORD, DYLAN R LAS VEGAS SPERM BANK 2870 S MARYLAND PKWY STE 250 LAS VEGAS, NV 89109-1548

COMMENTS: FASTING:UNKNOWN

Cytogenetic Report

CHROMOSOME ANALYSIS, BLOOD - 14596
Lab:EZ
CHROMOSOME ANALYSIS, BLOOD

Order ID: 25-38548
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: 20
Band Level: 450
Cells Analyzed: 5
Cells Karyotyped: 5

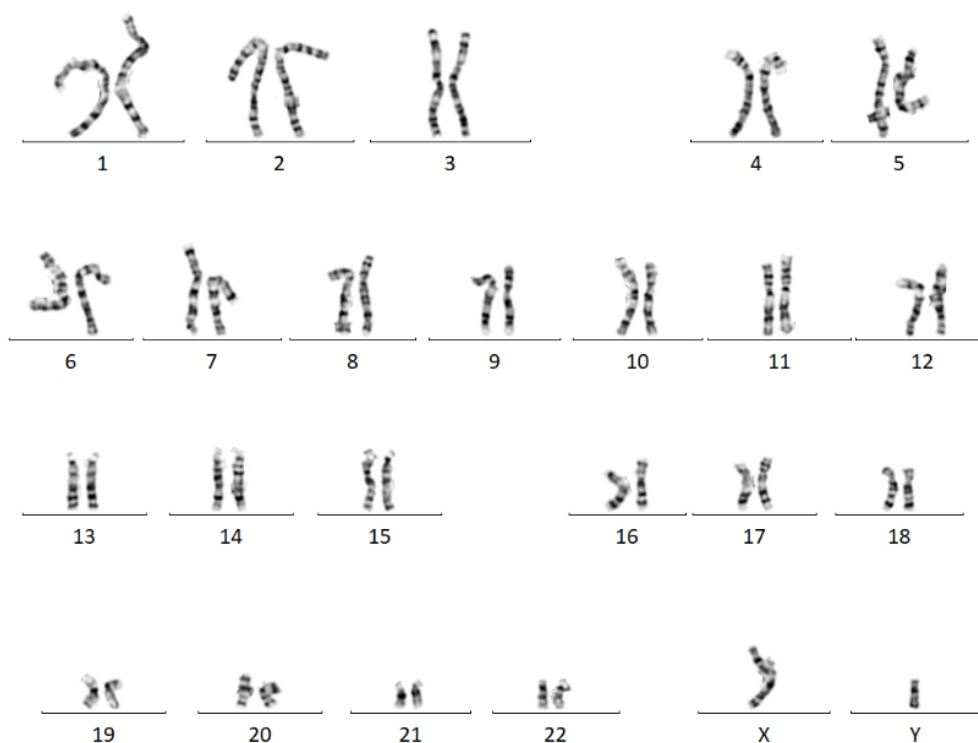
This test does not address genetic disorders that cannot be detected by standard cytogenetic methods or rare events such as low level mosaicism or subtle rearrangements. A portion of the testing was performed at SJC49

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

Electronic Signature: 1/30/2025 10:53 AM



Patient Information	Specimen Information	Client Information
20053, DONOR DOB: ████████ AGE: ██████ Gender: M Fasting: U Patient ID: 20053	Specimen: LV393354Y Collected: 01/22/2025 / 14:11 PST Received: 01/23/2025 / 02:00 PST Reported: 01/30/2025 / 08:35 PST	Client #: 88807473 FOORD, DYLAN R



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