



Donor 7110

Genetic Testing Summary

Fairfax Cryobank recommends reviewing this genetic testing summary with your healthcare provider to determine suitability.

Last Updated: 7/1/2025

Donor Reported Ancestry: Ukrainian, Greek

Jewish Ancestry: No

| Genetic Test* | Result | Comments/ Donor's Residual Risk** |
|---|--|---|
| Chromosome analysis (karyotype) | Normal male karyotype | No evidence of clinically significant chromosome abnormalities |
| Hemoglobin evaluation | Normal hemoglobin fractionation and MCV/MCH results | Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies |
| Expanded Genetic Disease Carrier Screening Panel attached- 514 diseases by gene sequencing. | <p>Carrier: AIPL1-related conditions (AIPL1)</p> <p>Carrier: SLC26A2-related conditions (SLC26A2)</p> <p>Negative for other genes sequenced.</p> | <p>Partner testing recommended before using this donor.</p> <p>Residual risks for negative results can be seen here:</p> <p>https://www.invitae.com/carrier-residual-risks/</p> |
| Special Testing | | |
| Genes: ABCC6, SH3TC2, MFN2, VWF | Negative via sequencing with deletion and duplication analysis | |

*No single test can screen for all genetic disorders. A negative screening result significantly reduces, but cannot eliminate, the risk for these conditions in a pregnancy.

**Donor residual risk is the chance the donor is still a carrier after testing negative.

| | | | | | |
|-------------------------------|------------|--------------------------------|-------------|-----------------------|-------------|
| Patient name: | 7110 Donor | Sample type: | Blood | Report date: | 04-MAY-2023 |
| DOB: | ██████████ | Sample collection date: | 08-FEB-2023 | Invitae #: | ██████████ |
| Sex assigned at birth: | Male | Sample accession date: | 09-FEB-2023 | Clinical team: | ██████████ |
| Gender: | | | | | ██████████ |
| Patient ID (MRN): | | | | | |

Reason for testing

Gamete donor

Test performed

Invitae Comprehensive Carrier Screen without X-linked Disorders

- Primary Panel (CF, SMA)
- Add-on Comprehensive Carrier Screen without X-linked Disorders genes

RE-REQUISITION REPORT: This report supersedes RQ4663162 (16-FEB-2023) and includes additional analyses.


RESULT: POSITIVE

This carrier test evaluated 514 gene(s) for genetic changes (variants) that are associated with an increased risk of having a child with a genetic condition. Knowledge of carrier status for one of these conditions may provide information that can be used to assist with family planning and/or preparation. Carrier screening is not intended for diagnostic purposes. To identify a potential genetic basis for a condition in the individual being tested, diagnostic testing for the gene(s) of interest is recommended.

This test shows the presence of clinically significant genetic change(s) in this individual in the gene(s) indicated below. No other clinically significant changes were identified in the remaining genes evaluated with this test.

| RESULTS | GENE | VARIANT(S) | INHERITANCE | PARTNER TESTING RECOMMENDED |
|--|---------|------------------------|---------------------|-----------------------------|
| Carrier: AIPL1-related conditions | AIPL1 | c.834G>A (p.Trp278*) | Autosomal recessive | Yes |
| Carrier: SLC26A2-related conditions | SLC26A2 | c.835C>T (p.Arg279Trp) | Autosomal recessive | Yes |



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DOB: [REDACTED]

Invitae #: [REDACTED]

Next steps

- See the table above for recommendations regarding testing of this individual's reproductive partner.
- Even for genes that have a negative test result, there is always a small risk that an individual could still be a carrier. This is called “residual risk.” See the Carrier detection rates and residual risks document.
- Discussion with a physician and/or genetic counselor is recommended to further review the implications of this test result and to understand these results in the context of any family history of a genetic condition.
- All patients, regardless of result, may wish to consider additional screening for hemoglobinopathies by complete blood count (CBC) and hemoglobin electrophoresis, if this has not already been completed.
- Individuals can register their tests at <https://www.invitae.com/patients/> to access online results, educational resources, and next steps.

Clinical summary



RESULT: CARRIER

AIPL1-related conditions

A single Pathogenic variant, c.834G>A (p.Trp278*), was identified in AIPL1.

What are AIPL1-related conditions?

AIPL1-related conditions include Leber congenital amaurosis (LCA), retinitis pigmentosa (RP), and cone-rod dystrophy (CRD). These conditions are retinal dystrophies, a class of inherited eye conditions characterized by degeneration of the rods and cones (photoreceptors) which are the cells in the retina that respond to light, as well as degeneration of the layer of tissue beneath the photoreceptors (retinal pigment epithelium [RPE]). Each of these conditions can be caused by changes in several different genes.

LCA typically causes severe visual impairment during infancy or early childhood, which is generally stable or very slowly progressive. The oculo-digital sign, which is a behavior consisting of eye poking, rubbing, and pressing, is also characteristic of LCA. A variety of other eye-related abnormalities, including involuntary eye movements (nystagmus), thinning and bulging of the clear covering at the front of the eye (cornea) [keratoconus], extreme farsightedness (hyperopia), little to no response by the pupils to light, and increased sensitivity to light (photophobia) may also occur. Electroretinography (ERG), an eye examination measuring visual function, typically detects little, if any, activity in the retina during infancy. Some individuals with LCA may also have intellectual disability.

The first symptom of RP is often difficulty seeing in low light settings (night blindness), which usually occurs during childhood or adolescence. Vision loss continues over years or decades and typically progresses to a loss of side (peripheral) vision, causing tunnel vision. Ultimately, central vision loss occurs.

Symptoms of CRD typically begin in childhood or adolescence and include reduced visual acuity (farsightedness or nearsightedness), loss of color perception, and photophobia. Symptoms may worsen over time to include night blindness and peripheral vision loss. Some affected individuals develop nystagmus.

In RP and CRD, intelligence and life expectancy are not typically affected. Most individuals with AIPL1-related retinal dystrophy have severe vision loss in childhood; however, in a small minority of cases, the vision loss may be mild and/or may not occur until adulthood.

Early initiation of medical, educational, and social services is recommended for affected individuals to maximize outcomes.

Next steps

Carrier testing for the reproductive partner is recommended.



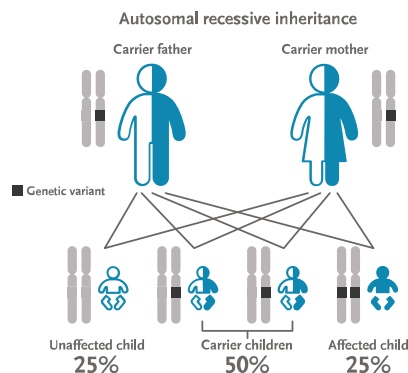
If your partner tests positive:

In autosomal recessive inheritance, an individual must have disease-causing genetic changes in each copy of the AIPL1 gene to be affected. Carriers, who have a disease-causing genetic change in only one copy of the gene, typically do not have symptoms. When both reproductive partners are carriers of an autosomal recessive condition, there is a 25% chance for each child to have the condition.



If your partner tests negative:

A negative carrier test result reduces, but does not eliminate, the chance that a person may be a carrier. The risk that a person could still be a carrier, even after a negative test result, is called a residual risk. See the table below for your partner's hypothetical residual risk after testing negative for AIPL1-related conditions. These values are provided only as a guide, are based on the detection rate for the





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condition as tested at Invitae, and assume a negative family history, the absence of symptoms, and vary based on the ethnic background of an individual. For genes associated with both dominant and recessive inheritance, the numbers provided apply to the recessive condition(s) associated with the gene.

| DISORDER (INHERITANCE) | GENE | ETHNICITY | CARRIER FREQUENCY BEFORE SCREENING | CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT |
|--|---------|------------|------------------------------------|---|
| AIPL1-related conditions (AR) NM_014336.4 | AIPL1 * | Pan-ethnic | 1 in 408 | 1 in 40700 |


RESULT: CARRIER

SLC26A2-related conditions

A single Pathogenic variant, c.835C>T (p.Arg279Trp), was identified in SLC26A2.

What are SLC26A2-related conditions?

SLC26A2-related conditions, also called sulfate transporter-related osteochondrodysplasias, include atelosteogenesis type 2 (AO2), achondrogenesis type 1B (ACG1B), diastrophic dysplasia (DTD), and multiple epiphyseal dysplasia type 4 (EDM4, or rMED). These conditions affect cartilage and bones and vary widely in severity. Symptoms of AO2 and ACG1B are severe, characterized by extremely short arms and legs, a narrow chest, and a prominent, rounded abdomen. Most affected infants are stillborn or die shortly after birth from respiratory failure. Individuals with DTD are short, with shortened limbs and a typical "hitchhiker" thumb. While some affected individuals may die in infancy, typically from respiratory complications, most live into adulthood. Individuals with EDM4 are more mildly affected. Symptoms typically include joint pain, abnormalities of the hands, feet, and knees, as well as side-to-side curvature of the spine (scoliosis). Height is usually within or near the normal range, and affected individuals live into adulthood. For DTD and EDM4, intelligence is not typically affected. Treatment may include physical therapy, orthopedics, and surgery. Follow-up depends on each affected individual's specific situation, and discussion with a healthcare provider should be considered.

Next steps

Carrier testing for the reproductive partner is recommended.



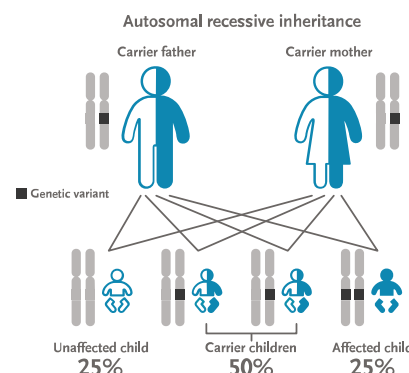
If your partner tests positive:

In autosomal recessive inheritance, an individual must have disease-causing genetic changes in each copy of the SLC26A2 gene to be affected. Carriers, who have a disease-causing genetic change in only one copy of the gene, typically do not have symptoms. When both reproductive partners are carriers of an autosomal recessive condition, there is a 25% chance for each child to have the condition.



If your partner tests negative:

A negative carrier test result reduces, but does not eliminate, the chance that a person may be a carrier. The risk that a person could still be a carrier, even after a negative test result, is called a residual risk. See the table below for your partner's hypothetical residual risk after testing negative for SLC26A2-related conditions. These values are provided only as a guide, are based on the detection rate for the condition as tested at Invitae, and assume a negative family history, the absence of symptoms, and vary based on the ethnic background of an individual. For genes associated with both dominant and recessive inheritance, the numbers provided apply to the recessive condition(s) associated with the gene.



| DISORDER (INHERITANCE) | GENE | ETHNICITY | CARRIER FREQUENCY BEFORE SCREENING | CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT |
|--|---------|------------|------------------------------------|---|
| SLC26A2-related conditions (AR) NM_000112.3 | SLC26A2 | Pan-ethnic | 1 in 158 | 1 in 3140 |

Results to note

ABCA4

- c.5603A>T (p.Asn1868Ile) was identified in the ABCA4 gene.
- This benign variant is not known to cause disease and does not impact this individual's risk to be a carrier for ABCA4-related conditions. Carrier testing for the reproductive partner is not indicated based on this result. See Variant details for more information.

SMN1

- Negative result. SMN1: 2 copies; c.*3+80T>G not detected.

Pseudodeficiency allele(s)

- Benign changes, c.1685T>C (p.Ile562Thr), known to be pseudodeficiency alleles, identified in the GALC gene. Pseudodeficiency alleles are not known to be associated with disease, including Krabbe disease.
- The presence of a pseudodeficiency allele does not impact this individual's risk to be a carrier. Individuals with pseudodeficiency alleles may exhibit false positive results on related biochemical tests, including newborn screening. However, pseudodeficiency alleles are not known to cause disease, even when there are two copies of the variant (homozygous) or when in combination with another disease-causing variant (compound heterozygous). Carrier testing for the reproductive partner is not indicated based on this result.

Variant details

ABCA4, Exon 40, c.5603A>T (p.Asn1868Ile), heterozygous, Benign (reportable variant)

- This sequence change replaces asparagine, which is neutral and polar, with isoleucine, which is neutral and non-polar, at codon 1868 of the ABCA4 protein (p.Asn1868Ile).
- This variant is present in population databases (rs1801466, gnomAD 7%), including several hundred presumably unaffected homozygous individuals.
- This missense change has been observed in individual(s) with late onset Stargardt disease with foveal sparing. However, the vast majority (estimated 95%) of homozygous and compound heterozygous individuals remain unaffected with penetrance ranging from 0.24% to 9.54% across published studies. This variant may modify disease severity and/or age of onset when it is present in combination with additional known pathogenic variants (e.g., when this variant is on the same chromosome as one or more deleterious variants, such as c.2588G>C, c.5461-10T>C, c.4496G>A, and/or c.2564G>A, and also on the opposite chromosome with a pathogenic variant). In other cases, disease progression is not impacted when this variant is one component of other complex alleles, such as with c.769-784C>T (PMID: 11328725, 28446513, 29971439, 30204727, 30480704, 30670881, 31614660, 31618761, 31884623, 32037395, 32307445, 32815999, 34440414, 34874912).
- ClinVar contains an entry for this variant (Variation ID: 99390).
- Advanced modeling of protein sequence and biophysical properties (such as structural, functional, and spatial information, amino acid conservation, physicochemical variation, residue mobility, and thermodynamic stability) performed at Invitae indicates that this missense variant is expected to disrupt ABCA4 protein function.
- Experimental studies are conflicting or provide insufficient evidence to determine the effect of this variant on ABCA4 function (PMID: 11017087, 32845050, 33375396).
- For these reasons, this variant has been classified as a Benign reportable variant.

AIPL1, Exon 6, c.834G>A (p.Trp278*), heterozygous, PATHOGENIC

- This sequence change creates a premature translational stop signal (p.Trp278*) in the AIPL1 gene. While this is not anticipated to result in nonsense mediated decay, it is expected to disrupt the last 107 amino acid(s) of the AIPL1 protein.
- This variant is present in population databases (rs62637014, gnomAD 0.06%).



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- This premature translational stop signal has been observed in individual(s) with Leber congenital amaurosis (PMID: 10615133, 10873396, 15249368, 21474771, 22412862). It has also been observed to segregate with disease in related individuals.
- ClinVar contains an entry for this variant (Variation ID: 5565).
- Algorithms developed to predict the effect of variants on protein structure and function are not available or were not evaluated for this variant.
- Experimental studies have shown that this premature translational stop signal affects AIPL1 function (PMID: 15347646, 25799540).
- For these reasons, this variant has been classified as Pathogenic.

SLC26A2, Exon 3, c.835C>T (p.Arg279Trp), heterozygous, PATHOGENIC

- This sequence change replaces arginine, which is basic and polar, with tryptophan, which is neutral and slightly polar, at codon 279 of the SLC26A2 protein (p.Arg279Trp).
- This variant is present in population databases (rs104893915, gnomAD 0.2%), and has an allele count higher than expected for a pathogenic variant.
- This missense change has been observed in individuals with SLC26A2-related disease (PMID: 8571951, 9342225, 10465113, 10482955, 16642506, 21077202, 22052783, 23840040, 27065010).
- ClinVar contains an entry for this variant (Variation ID: 4089).
- Advanced modeling of protein sequence and biophysical properties (such as structural, functional, and spatial information, amino acid conservation, physicochemical variation, residue mobility, and thermodynamic stability) performed at Invitae indicates that this missense variant is expected to disrupt SLC26A2 protein function.
- Experimental studies have shown that this missense change affects SLC26A2 function (PMID: 15294877, 20219950).
- For these reasons, this variant has been classified as Pathogenic.

Residual risk

No carrier test can detect 100% of carriers. There still remains a small risk of being a carrier after a negative test (residual risk). Residual risk values assume a negative family history and are inferred from published carrier frequencies and estimated detection rates based on testing technologies used at Invitae. You can view Invitae's complete Carrier detection rates and residual risks document (containing all carrier genes) online at <https://www.invitae.com/carrier-residual-risks/>. Additionally, the order-specific information for this report is available to download in the portal (under this order's documents) or can be requested by contacting Invitae Client Services. The complete Carrier detection rates and residual risks document will not be applicable for any genes with specimen-specific limitations in sequencing and/or deletion/duplication coverage. Please see the final bullet point in the Limitations section of this report to view if this specimen had any gene-specific coverage gaps.

Genes analyzed

This table represents a complete list of genes analyzed for this individual, including the relevant gene transcript(s). If more than one transcript is listed for a single gene, variants were reported using the first transcript listed unless otherwise indicated in the report. An asterisk (*) indicates that this gene has a limitation. Please see the Limitations section for details. Results are negative, unless otherwise indicated in the report.

| GENE | TRANSCRIPT | GENE | TRANSCRIPT | GENE | TRANSCRIPT |
|---------|-------------|----------|-------------------------|----------|----------------|
| AAAS | NM_015665.5 | AP1S1 | NM_001283.3 | CBS | NM_000071.2 |
| ABCA12 | NM_173076.2 | AQP2 | NM_000486.5 | CC2D1A | NM_017721.5 |
| ABCA3 | NM_001089.2 | ARG1 | NM_000045.3 | CC2D2A | NM_001080522.2 |
| ABCA4 | NM_000350.2 | ARL6 | NM_177976.2 | CCDC103 | NM_213607.2 |
| ABCB11 | NM_003742.2 | ARSA | NM_000487.5 | CCDC39 | NM_181426.1 |
| ABCB4 | NM_000443.3 | ARSB | NM_000046.3 | CCDC88C | NM_001080414.3 |
| ABCC2* | NM_000392.4 | ASL | NM_000048.3 | CD3D | NM_000732.4 |
| ABCC8 | NM_000352.4 | ASNS | NM_133436.3 | CD3E | NM_000733.3 |
| ACAD9 | NM_014049.4 | ASPA | NM_000049.2 | CD40 | NM_001250.5 |
| ACADM | NM_000016.5 | ASS1 | NM_000050.4 | CD59 | NM_203330.2 |
| ACADVL | NM_000018.3 | ATM* | NM_000051.3 | CDH23 | NM_022124.5 |
| ACAT1 | NM_000019.3 | ATP6V1B1 | NM_001692.3 | CEP152 | NM_014985.3 |
| ACOX1 | NM_004035.6 | ATP7B | NM_000053.3 | CEP290 | NM_025114.3 |
| ACSF3 | NM_174917.4 | ATP8B1* | NM_005603.4 | CERKL | NM_001030311.2 |
| ADA | NM_000022.2 | BBS1 | NM_024649.4 | CFTR* | NM_000492.3 |
| ADAMTS2 | NM_014244.4 | BBS10 | NM_024685.3 | CHAT | NM_020549.4 |
| ADAMTS4 | NM_019032.5 | BBS12 | NM_152618.2 | CHRNE | NM_000080.3 |
| ADGRG1 | NM_005682.6 | BBS2 | NM_031885.3 | CHRNA | NM_005199.4 |
| ADGRV1 | NM_032119.3 | BBS4 | NM_033028.4 | CIITA | NM_000246.3 |
| AGA | NM_000027.3 | BBS5 | NM_152384.2 | CLCN1 | NM_000083.2 |
| AGL | NM_000642.2 | BBS7 | NM_176824.2 | CLN3 | NM_001042432.1 |
| AGPS | NM_003659.3 | BBS9* | NM_198428.2 | CLN5 | NM_006493.2 |
| AGXT | NM_000030.2 | BCKDHA | NM_000709.3 | CLN6 | NM_017882.2 |
| AHI1 | NM_017651.4 | BCKDHB | NM_183050.2 | CLN8 | NM_018941.3 |
| AIPL1* | NM_014336.4 | BCS1L | NM_004328.4 | CLRN1 | NM_174878.2 |
| AIRE | NM_000383.3 | BLM | NM_000057.3 | CNGB3 | NM_019098.4 |
| ALDH3A2 | NM_000382.2 | BLOC1S3 | NM_212550.4 | COL11A2* | NM_080680.2 |
| ALDH7A1 | NM_001182.4 | BLOC1S6 | NM_012388.3 | COL17A1 | NM_000494.3 |
| ALDOB | NM_000035.3 | BMP1 | NM_006129.4;NM_001199.3 | COL27A1 | NM_032888.3 |
| ALG1 | NM_019109.4 | BRIP1 | NM_032043.2 | COL4A3 | NM_000091.4 |
| ALG6 | NM_013339.3 | BSND | NM_057176.2 | COL4A4 | NM_000092.4 |
| ALMS1 | NM_015120.4 | BTD | NM_000060.3 | COL7A1 | NM_000094.3 |
| ALPL | NM_000478.5 | CAD | NM_004341.4 | COX15 | NM_004376.6 |
| AMN* | NM_030943.3 | CANT1 | NM_138793.3 | CPS1 | NM_001875.4 |
| AMT | NM_000481.3 | CAPN3 | NM_000070.2 | CPT1A | NM_001876.3 |
| ANO10* | NM_018075.3 | CASQ2 | NM_001232.3 | CPT2 | NM_000098.2 |



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DOB: [REDACTED]

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| GENE | TRANSCRIPT |
|----------|----------------|
| CRB1 | NM_201253.2 |
| CRTAP | NM_006371.4 |
| CTNS | NM_004937.2 |
| CTSA | NM_000308.3 |
| CTSC | NM_001814.5 |
| CTSD | NM_001909.4 |
| CTSK | NM_000396.3 |
| CYBA | NM_000101.3 |
| CYP11A1 | NM_000781.2 |
| CYP11B1 | NM_000497.3 |
| CYP11B2 | NM_000498.3 |
| CYP17A1 | NM_000102.3 |
| CYP19A1 | NM_031226.2 |
| CYP1B1 | NM_000104.3 |
| CYP21A2* | NM_000500.7 |
| CYP27A1 | NM_000784.3 |
| CYP27B1 | NM_000785.3 |
| CYP7B1 | NM_004820.3 |
| DBT | NM_001918.3 |
| DCAF17 | NM_025000.3 |
| DCLRE1C | NM_001033855.2 |
| DDX11* | NM_030653.3 |
| DFNB59 | NM_001042702.3 |
| DGAT1 | NM_012079.5 |
| DGUOK | NM_080916.2 |
| DHCR7 | NM_001360.2 |
| DHDDS | NM_024887.3 |
| DLD | NM_000108.4 |
| DLL3 | NM_016941.3 |
| DNAH11 | NM_001277115.1 |
| DNAH5 | NM_001369.2 |
| DNAI1 | NM_012144.3 |
| DNAI2 | NM_023036.4 |
| DNMT3B | NM_006892.3 |
| DOK7 | NM_173660.4 |
| DUOX2* | NM_014080.4 |
| DYNC2H1 | NM_001080463.1 |
| DYSF | NM_003494.3 |
| EIF2AK3 | NM_004836.6 |

| GENE | TRANSCRIPT |
|---------|----------------|
| EIF2B1 | NM_001414.3 |
| EIF2B2 | NM_014239.3 |
| EIF2B3 | NM_020365.4 |
| EIF2B4 | NM_015636.3 |
| EIF2B5 | NM_003907.2 |
| ELP1 | NM_003640.3 |
| EPG5 | NM_020964.2 |
| ERCC2 | NM_000400.3 |
| ERCC6 | NM_000124.3 |
| ERCC8 | NM_000082.3 |
| ESCO2 | NM_001017420.2 |
| ETFA | NM_000126.3 |
| ETFB | NM_001985.2 |
| ETFDH | NM_004453.3 |
| ETHE1 | NM_014297.3 |
| EVC | NM_153717.2 |
| EVC2 | NM_147127.4 |
| EXOSC3 | NM_016042.3 |
| EYS* | NM_001142800.1 |
| FAH* | NM_000137.2 |
| FAM161A | NM_001201543.1 |
| FANCA | NM_000135.2 |
| FANCC | NM_000136.2 |
| FANCD2* | NM_033084.3 |
| FANCE | NM_021922.2 |
| FANCG | NM_004629.1 |
| FANCI | NM_001113378.1 |
| FANCL* | NM_018062.3 |
| FBP1 | NM_000507.3 |
| FBXO7 | NM_012179.3 |
| FH* | NM_000143.3 |
| FKBP10 | NM_021939.3 |
| FKRP | NM_024301.4 |
| FKTN | NM_001079802.1 |
| FMO3 | NM_006894.6 |
| FOXN1 | NM_003593.2 |
| FOXRED1 | NM_017547.3 |
| FRAS1 | NM_025074.6 |
| FREM2 | NM_207361.5 |

| GENE | TRANSCRIPT |
|--------|----------------|
| FUCA1 | NM_000147.4 |
| G6PC | NM_000151.3 |
| G6PC3 | NM_138387.3 |
| GAA | NM_000152.3 |
| GALC* | NM_000153.3 |
| GALE* | NM_000403.3 |
| GALK1 | NM_000154.1 |
| GALNS | NM_000512.4 |
| GALNT3 | NM_004482.3 |
| GALT | NM_000155.3 |
| GAMT | NM_000156.5 |
| GATM | NM_001482.2 |
| GBA* | NM_001005741.2 |
| GBE1 | NM_000158.3 |
| GCDH | NM_000159.3 |
| GCH1 | NM_000161.2 |
| GDF5 | NM_000557.4 |
| GFM1 | NM_024996.5 |
| GHR* | NM_000163.4 |
| GJB2 | NM_004004.5 |
| GLB1 | NM_000404.2 |
| GLDC | NM_000170.2 |
| GLE1 | NM_001003722.1 |
| GNE* | NM_001128227.2 |
| GNPAT | NM_014236.3 |
| GNPTAB | NM_024312.4 |
| GNPTG | NM_032520.4 |
| GNS | NM_002076.3 |
| GORAB | NM_152281.2 |
| GRHPR | NM_012203.1 |
| GRIP1 | NM_021150.3 |
| GSS | NM_000178.2 |
| GUCY2D | NM_000180.3 |
| GUSB | NM_000181.3 |
| HADH | NM_005327.4 |
| HADHA | NM_000182.4 |
| HADHB | NM_000183.2 |
| HAMP | NM_021175.2 |
| HAX1 | NM_006118.3 |


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Invitae #: [REDACTED]

| GENE | TRANSCRIPT |
|---------|----------------|
| HBA1* | NM_000558.4 |
| HBA2 | NM_000517.4 |
| HBB | NM_000518.4 |
| HEXA | NM_000520.4 |
| HEXB | NM_000521.3 |
| HGSNAT | NM_152419.2 |
| HJV | NM_213653.3 |
| HLCS | NM_000411.6 |
| HMGCL | NM_000191.2 |
| HMOX1 | NM_002133.2 |
| HOGA1 | NM_138413.3 |
| HPD | NM_002150.2 |
| HPS1 | NM_000195.4 |
| HPS3 | NM_032383.4 |
| HPS4 | NM_022081.5 |
| HPS5 | NM_181507.1 |
| HPS6 | NM_024747.5 |
| HSD17B3 | NM_000197.1 |
| HSD17B4 | NM_000414.3 |
| HSD3B2 | NM_000198.3 |
| HYAL1 | NM_153281.1 |
| HYLS1 | NM_145014.2 |
| IDUA | NM_000203.4 |
| IGHMBP2 | NM_002180.2 |
| IKBB | NM_001556.2 |
| IL7R | NM_002185.3 |
| INVS | NM_014425.3 |
| ITGA6 | NM_000210.3 |
| ITGB3 | NM_000212.2 |
| ITGB4 | NM_001005731.2 |
| IVD | NM_002225.3 |
| JAK3 | NM_000215.3 |
| KCNJ1 | NM_000220.4 |
| KCNJ11 | NM_000525.3 |
| LAMA2 | NM_000426.3 |
| LAMA3 | NM_000227.4 |
| LAMB3 | NM_000228.2 |
| LAMC2 | NM_005562.2 |
| LARGE1 | NM_004737.4 |

| GENE | TRANSCRIPT |
|---------|----------------|
| LCA5 | NM_181714.3 |
| LDLR | NM_000527.4 |
| LDLRAP1 | NM_015627.2 |
| LHX3 | NM_014564.4 |
| LIFR* | NM_002310.5 |
| LIG4 | NM_002312.3 |
| LIPA | NM_000235.3 |
| LMBRD1 | NM_018368.3 |
| LOXHD1 | NM_144612.6 |
| LPL | NM_000237.2 |
| LRAT | NM_004744.4 |
| LRP2 | NM_004525.2 |
| LRPPRC | NM_133259.3 |
| LYST | NM_000081.3 |
| MAK | NM_001242957.2 |
| MAN2B1 | NM_000528.3 |
| MANBA | NM_005908.3 |
| MCEE | NM_032601.3 |
| MCOLN1 | NM_020533.2 |
| MCPH1 | NM_024596.4 |
| MECR | NM_016011.3 |
| MED17 | NM_004268.4 |
| MESP2 | NM_001039958.1 |
| MFSD8 | NM_152778.2 |
| MKKS | NM_018848.3 |
| MKS1 | NM_017777.3 |
| MLC1* | NM_015166.3 |
| MLYCD | NM_012213.2 |
| MMAA | NM_172250.2 |
| MMAB | NM_052845.3 |
| MMACHC | NM_015506.2 |
| MMADHC | NM_015702.2 |
| MOCS1 | NM_001358530.2 |
| MOCS2A | NM_176806.3 |
| MOCS2B | NM_004531.4 |
| MPI | NM_002435.2 |
| MPL | NM_005373.2 |
| MPV17 | NM_002437.4 |
| MRE11 | NM_005591.3 |

| GENE | TRANSCRIPT |
|---------|-------------------------|
| MTHFR* | NM_005957.4 |
| MTR | NM_000254.2 |
| MTRR | NM_002454.2 |
| MTTP | NM_000253.3 |
| MUSK | NM_005592.3 |
| MUT | NM_000255.3 |
| MVK | NM_000431.3 |
| MYO15A | NM_016239.3 |
| MYO7A | NM_000260.3 |
| NAGA | NM_000262.2 |
| NAGLU | NM_000263.3 |
| NAGS | NM_153006.2 |
| NBN | NM_002485.4 |
| NCF2 | NM_000433.3 |
| NDRG1 | NM_006096.3 |
| NDUFAF2 | NM_174889.4 |
| NDUFAF5 | NM_024120.4 |
| NDUFS4 | NM_002495.3 |
| NDUFS6 | NM_004553.4 |
| NDUFS7 | NM_024407.4 |
| NDUFV1 | NM_007103.3 |
| NEB* | NM_001271208.1 |
| NEU1 | NM_000434.3 |
| NGLY1 | NM_018297.3 |
| NPC1 | NM_000271.4 |
| NPC2 | NM_006432.3 |
| NPHP1 | NM_000272.3 |
| NPHS1 | NM_004646.3 |
| NPHS2 | NM_014625.3 |
| NR2E3 | NM_014249.3 |
| NSMCE3 | NM_138704.3 |
| NTRK1 | NM_001012331.1 |
| OAT* | NM_000274.3 |
| OCA2 | NM_000275.2 |
| OPA3 | NM_025136.3 |
| OSTM1 | NM_014028.3 |
| OTOA* | NM_144672.3 |
| OTOF | NM_194248.2;NM_194323.2 |
| P3H1 | NM_022356.3 |


Patient name: 7110 Donor

DOB: [REDACTED]

Invitae #: [REDACTED]

| GENE | TRANSCRIPT |
|---------|-----------------------------|
| PAH | NM_000277.1 |
| PANK2 | NM_153638.2 |
| PC | NM_000920.3 |
| PCBD1 | NM_000281.3 |
| PCCA | NM_000282.3 |
| PCCB | NM_000532.4 |
| PCDH15 | NM_033056.3 |
| PCNT | NM_006031.5 |
| PDHB | NM_000925.3 |
| PEPD | NM_000285.3 |
| PET100 | NM_001171155.1 |
| PEX1* | NM_000466.2 |
| PEX10 | NM_153818.1 |
| PEX12 | NM_000286.2 |
| PEX13 | NM_002618.3 |
| PEX16 | NM_004813.2 |
| PEX2 | NM_000318.2 |
| PEX26 | NM_017929.5 |
| PEX5 | NM_001131025.1 |
| PEX6 | NM_000287.3 |
| PEX7 | NM_000288.3 |
| PFKM | NM_000289.5 |
| PGM3 | NM_001199917.1 |
| PHGDH | NM_006623.3 |
| PHKB | NM_000293.2;NM_00103183.5.2 |
| PHKG2 | NM_000294.2 |
| PHYH | NM_006214.3 |
| PIGN | NM_176787.4 |
| PKHD1* | NM_138694.3 |
| PLA2G6 | NM_003560.2 |
| PLEKHG5 | NM_020631.4 |
| PLOD1 | NM_000302.3 |
| PMM2 | NM_000303.2 |
| PNPO | NM_018129.3 |
| POLG | NM_002693.2 |
| POLH | NM_006502.2 |
| POMGNT1 | NM_017739.3 |
| POMT1 | NM_007171.3 |
| POMT2 | NM_013382.5 |

| GENE | TRANSCRIPT |
|----------|----------------|
| POR | NM_000941.2 |
| POU1F1 | NM_000306.3 |
| PPT1 | NM_000310.3 |
| PRCD | NM_001077620.2 |
| PRDM5 | NM_018699.3 |
| PRF1 | NM_001083116.1 |
| PROP1 | NM_006261.4 |
| PSAP | NM_002778.3 |
| PTPRC* | NM_002838.4 |
| PTS | NM_000317.2 |
| PUS1 | NM_025215.5 |
| PYGM | NM_005609.3 |
| QDPR | NM_000320.2 |
| RAB23 | NM_183227.2 |
| RAG1 | NM_000448.2 |
| RAG2 | NM_000536.3 |
| RAPSN | NM_005055.4 |
| RARS2 | NM_020320.3 |
| RDH12 | NM_152443.2 |
| RLBP1 | NM_000326.4 |
| RMRP | NR_003051.3 |
| RNASEH2A | NM_006397.2 |
| RNASEH2B | NM_024570.3 |
| RNASEH2C | NM_032193.3 |
| RPE65 | NM_000329.2 |
| RPGRIP1L | NM_015272.2 |
| RTEL1 | NM_001283009.1 |
| RXYLT1 | NM_014254.2 |
| RYR1 | NM_000540.2 |
| SACS | NM_014363.5 |
| SAMD9 | NM_017654.3 |
| SAMHD1 | NM_015474.3 |
| SCO2 | NM_005138.2 |
| SEC23B | NM_006363.4 |
| SEPSECS | NM_016955.3 |
| SGCA | NM_000023.2 |
| SGCB | NM_000232.4 |
| SGCD | NM_000337.5 |
| SGCG | NM_000231.2 |

| GENE | TRANSCRIPT |
|----------|----------------|
| SGSH | NM_000199.3 |
| SKIV2L | NM_006929.4 |
| SLC12A1 | NM_000338.2 |
| SLC12A3 | NM_000339.2 |
| SLC12A6 | NM_133647.1 |
| SLC17A5 | NM_012434.4 |
| SLC19A2 | NM_006996.2 |
| SLC19A3 | NM_025243.3 |
| SLC1A4 | NM_003038.4 |
| SLC22A5 | NM_003060.3 |
| SLC25A13 | NM_014251.2 |
| SLC25A15 | NM_014252.3 |
| SLC25A20 | NM_000387.5 |
| SLC26A2 | NM_000112.3 |
| SLC26A3 | NM_000111.2 |
| SLC26A4 | NM_000441.1 |
| SLC27A4 | NM_005094.3 |
| SLC35A3 | NM_012243.2 |
| SLC37A4 | NM_001164277.1 |
| SLC38A8 | NM_001080442.2 |
| SLC39A4 | NM_130849.3 |
| SLC45A2 | NM_016180.4 |
| SLC4A11 | NM_032034.3 |
| SLC5A5 | NM_000453.2 |
| SLC7A7 | NM_001126106.2 |
| SMARCA1 | NM_014140.3 |
| SMN1* | NM_000344.3 |
| SMPD1 | NM_000543.4 |
| SNAP29 | NM_004782.3 |
| SPG11 | NM_025137.3 |
| SPR | NM_003124.4 |
| SRD5A2 | NM_000348.3 |
| ST3GAL5 | NM_003896.3 |
| STAR | NM_000349.2 |
| STX11 | NM_003764.3 |
| STXBP2 | NM_006949.3 |
| SUMF1 | NM_182760.3 |
| SUOX | NM_000456.2 |
| SURF1 | NM_003172.3 |


Patient name: 7110 Donor

DOB: [REDACTED]

Invitae #: [REDACTED]

| GENE | TRANSCRIPT |
|---------|----------------|
| SYNE4 | NM_001039876.2 |
| TANGO2 | NM_152906.6 |
| TAT | NM_000353.2 |
| TBCD | NM_005993.4 |
| TBCE* | NM_003193.4 |
| TCIRG1 | NM_006019.3 |
| TCN2 | NM_000355.3 |
| TECPR2 | NM_014844.3 |
| TERT | NM_198253.2 |
| TF | NM_001063.3 |
| TFR2 | NM_003227.3 |
| TG* | NM_003235.4 |
| TGM1 | NM_000359.2 |
| TH | NM_199292.2 |
| TK2 | NM_004614.4 |
| TMC1 | NM_138691.2 |
| TMEM216 | NM_001173990.2 |
| TMEM67 | NM_153704.5 |
| TMPRSS3 | NM_024022.2 |
| TPO | NM_000547.5 |
| TPP1 | NM_000391.3 |
| TREX1 | NM_033629.4 |
| TRIM32 | NM_012210.3 |
| TRIM37 | NM_015294.4 |
| TRMU | NM_018006.4 |
| TSEN54 | NM_207346.2 |
| TSFM* | NM_001172696.1 |
| TSHB | NM_000549.4 |
| TSHR | NM_000369.2 |
| TTC37 | NM_014639.3 |
| TTPA | NM_000370.3 |
| TULP1 | NM_003322.4 |
| TYMP | NM_001953.4 |
| TYR* | NM_000372.4 |
| TYRP1 | NM_000550.2 |
| UBR1 | NM_174916.2 |
| UNC13D | NM_199242.2 |
| USH1C* | NM_005709.3 |
| USH2A | NM_206933.2 |

| GENE | TRANSCRIPT |
|---------|----------------|
| VDR | NM_001017535.1 |
| VLDLR | NM_003383.4 |
| VPS11 | NM_021729.5 |
| VPS13A* | NM_033305.2 |
| VPS13B | NM_017890.4 |
| VPS45 | NM_007259.4 |
| VPS53* | NM_001128159.2 |
| VRK1 | NM_003384.2 |
| VSX2 | NM_182894.2 |
| WISP3 | NM_003880.3 |
| WNT10A | NM_025216.2 |
| WRN* | NM_000553.4 |
| XPA | NM_000380.3 |
| XPC | NM_004628.4 |
| ZBTB24 | NM_014797.2 |
| ZFYVE26 | NM_015346.3 |
| ZNF469 | NM_001127464.2 |

Methods

- Genomic DNA obtained from the submitted sample is enriched for targeted regions using a hybridization-based protocol, and sequenced using Illumina technology. Unless otherwise indicated, all targeted regions are sequenced with $\geq 50\times$ depth or are supplemented with additional analysis. Reads are aligned to a reference sequence (GRCh37), and sequence changes are identified and interpreted in the context of a single clinically relevant transcript, indicated in the Genes Analyzed table. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 20bp of flanking intronic sequence, and other specific genomic regions demonstrated to be causative of disease at the time of assay design. Promoters, untranslated regions, and other non-coding regions are not otherwise interrogated. Exonic deletions and duplications are called using an in-house algorithm that determines copy number at each target by comparing the read depth for each target in the proband sequence with both mean read-depth and read-depth distribution, obtained from a set of clinical samples. Markers across the X and Y chromosomes are analyzed for quality control purposes and may detect deviations from the expected sex chromosome complement. Such deviations may be included in the report in accordance with internal guidelines. Invitae utilizes a classification methodology to identify next-generation sequencing (NGS)-detected variants that require orthogonal confirmation (Lincoln, et al. J Mol Diagn. 2019 Mar;21(2):318-329). Confirmation of the presence and location of reportable variants is performed as needed based on stringent criteria using one of several validated orthogonal approaches (PubMed ID 30610921). Sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778). Confirmatory sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778).

The following additional analyses are performed if relevant to the requisition. For GBA the reference genome has been modified to mask the sites of polymorphic paralog sequence variants (PSVs) in both the gene and pseudogene. For CYP21A2 and GBA, if one or more reportable variants, gene conversion, or fusion event is identified via our NGS pipeline (see Limitations), these variants are confirmed by PacBio sequencing of an amplicon generated by long-range PCR and subsequent short-range PCR. In some cases, it may not be possible to disambiguate between the gene and pseudogene. For GJB2, the reportable range includes large upstream deletions overlapping GJB6. For HBA1/2, the reference genome has been modified to force some sequencing reads derived from HBA1 to align to HBA2, and variant calling algorithms are modified to support an expectation of 4 alleles in these regions. HBA1/2 copy number calling is performed by a custom hypothesis testing algorithm which generates diplotype calls. If sequence data for a sample does not support a unique high confidence match from among hypotheses tested, that sample is flagged for manual review. Copy number variation is only reported for coding sequence of HBA1 and HBA2 and the HS-40 region. This assay does not distinguish among the $\alpha 3.7$ subtypes, and all $\alpha 3.7$ variants are called as HBA1 deletions. This assay may not detect overlapping copy gain and copy loss events when the breakpoints of those events are similar. For FMR1, cytosine-guanine-guanine (CGG) triplet repeats in the 5' untranslated region (5' UTR) of the FMR1 gene are detected by triplet repeat-primed PCR (RP-PCR) with fluorescently labeled primers followed by capillary electrophoresis. Reference ranges: Normal: <45 CGG repeats, intermediate: 45-54 CGG repeats, premutation: 55-200 CGG repeats, full mutation: >200 CGG repeats. For alleles with 55-90 triplet repeats, the region surrounding the FMR1 repeat is amplified by PCR. The PCR amplicons are then processed through PacBio SMRTBell library prep and sequenced using PacBio long read technology. The number of AGG interruptions within the 55-90 triplet repeat is read directly from the resulting DNA sequences.

- This report only includes variants that have a clinically significant association with the conditions tested as of the report date. Variants of uncertain significance, benign variants, and likely benign variants are not included in this report. However, if additional evidence becomes available to indicate that the clinical significance of a variant has changed, Invitae may update this report and provide notification.
- A PMID is a unique identifier referring to a published, scientific paper. Search by PMID at <http://www.ncbi.nlm.nih.gov/pubmed>.
- An rsID is a unique identifier referring to a single genomic position, and is used to associate population frequency information with sequence changes at that position. Reported population frequencies are derived from a number of public sites that aggregate data from large-scale population sequencing projects, including ExAC (<http://exac.broadinstitute.org>), gnomAD (<http://gnomad.broadinstitute.org>), and dbSNP (<http://ncbi.nlm.nih.gov/SNP>).

Disclaimer

DNA studies do not constitute a definitive test for the selected condition(s) in all individuals. It should be realized that there are possible sources of error. Errors can result from trace contamination, rare technical errors, rare genetic variants that interfere with analysis, recent scientific developments, and alternative classification systems. This test should be one of many aspects used by the healthcare provider to help with a diagnosis and treatment plan, but it is not a diagnosis itself. This test was developed and its performance characteristics determined by Invitae. It has not been cleared or approved by

the FDA. The laboratory is regulated under the Clinical Laboratory Improvement Act (CLIA) as qualified to perform high-complexity clinical tests (CLIA ID: 05D2040778). This test is used for clinical purposes. It should not be regarded as investigational or for research.

Limitations

- Based on validation study results, this assay achieves >99% analytical sensitivity and specificity for single nucleotide variants, insertions and deletions <15bp in length, and exon-level deletions and duplications. Invitae's methods also detect insertions and deletions larger than 15bp but smaller than a full exon but sensitivity for these may be marginally reduced. Invitae's deletion/duplication analysis determines copy number at a single exon resolution at virtually all targeted exons. However, in rare situations, single-exon copy number events may not be analyzed due to inherent sequence properties or isolated reduction in data quality. Certain types of variants, such as structural rearrangements (e.g. inversions, gene conversion events, translocations, etc.) or variants embedded in sequence with complex architecture (e.g. short tandem repeats or segmental duplications), may not be detected. Additionally, it may not be possible to fully resolve certain details about variants, such as mosaicism, phasing, or mapping ambiguity. Unless explicitly guaranteed, sequence changes in the promoter, non-coding exons, and other non-coding regions are not covered by this assay. Please consult the test definition on our website for details regarding regions or types of variants that are covered or excluded for this test. This report reflects the analysis of an extracted genomic DNA sample. While this test is intended to reflect the analysis of extracted genomic DNA from a referred patient, in very rare cases the analyzed DNA may not represent that individual's constitutional genome, such as in the case of a circulating hematolymphoid neoplasm, bone marrow transplant, blood transfusion, chimerism, culture artifact or maternal cell contamination.
- FANCL:** Sequencing analysis for exons 4, 10 includes only cds +/- 10 bp. **VPS53:** Sequencing analysis for exons 14 includes only cds +/- 5 bp. **FH:** Sequencing analysis for exons 9 includes only cds +/- 10 bp. **GALC:** Deletion/duplication analysis is not offered for exon 6. **GNE:** Sequencing analysis for exons 8 includes only cds +/- 10 bp. **AIPL1:** Sequencing analysis for exons 2 includes only cds +/- 10 bp. **TG:** Deletion/duplication analysis is not offered for exon 18. Sequencing analysis for exons 44 includes only cds +/- 0 bp. **EYS:** Sequencing analysis for exons 30 includes only cds +/- 0 bp. **HBA1/2:** This assay is designed to detect deletions and duplications of HBA1 and/or HBA2, resulting from the -alpha20.5, --MED, --SEA, --FIL/--THAI, -alpha3.7, -alpha4.2, anti3.7 and anti4.2. Sensitivity to detect other copy number variants may be reduced. Detection of overlapping deletion and duplication events will be limited to combinations of events with significantly differing boundaries. In addition, deletion of the enhancer element HS-40 and the sequence variant, Constant Spring (NM_000517.4:c.427T>C), can be identified by this assay. **GHR:** Deletion/duplication and sequencing analysis is not offered for exon 3. **CYP21A2:** Analysis includes the most common variants (c.92C>T(p.Pro31Leu), c.293-13C>G (intronic), c.332_339delGAGACTAC (p.Gly111Valfs*21), c.518T>A (p.Ile173Asn), c.710T>A (p.Ile237Asn), c.713T>A (p.Val238Glu), c.719T>A (p.Met240Lys), c.844G>T (p.Val282Leu), c.923dupT (p.Leu308Phefs*6), c.955C>T (p.Gln319*), c.1069C>T(p.Arg357Trp), c.1360C>T (p.Pro454Ser) and the 30Kb deletion) as well as select rare HGMD variants only (list available upon request). Full gene duplications are reported only in the presence of a pathogenic variant(s). When a duplication and a pathogenic variant(s) is identified, phase (cis/trans) cannot be determined. Full gene deletion analysis is not offered. Sensitivity to detect these variants, if they result from complex gene conversion/fusion events, may be reduced. **ABCC2:** Deletion/duplication analysis is not offered for exons 24-25. **FAH:** Deletion/duplication analysis is not offered for exon 14. **LIFR:** Sequencing analysis for exons 3 includes only cds +/- 5 bp. **MLC1:** Sequencing analysis for exons 11 includes only cds +/- 10 bp. **MTHFR:** The NM_005957.4:c.665C>T (p.Ala222Val) (aka 677C>T) and c.1286A>C (p.Glu429Ala) (aka 1298A>C) variants are not reported in our primary report. **NEB:** Deletion/duplication analysis is not offered for exons 82-105. NEB variants in this region with no evidence towards pathogenicity are not included in this report, but are available upon request. **PEX1:** Sequencing analysis for exons 16 includes only cds +/- 0 bp. **PKHD1:** Deletion/duplication analysis is not offered for exon 13. **USH1C:** Deletion/duplication analysis is not offered for exons 5-6. **BBS9:** Deletion/duplication analysis is not offered for exon 4. **WRN:** Deletion/duplication analysis is not offered for exons 10-11. Sequencing analysis for exons 8, 10-11 includes only cds +/- 10 bp. **GALE:** Sequencing analysis for exons 10 includes only cds +/- 5 bp. **OTOA:** Deletion/duplication and sequencing analysis is not offered for exons 20-28. **DUOX2:** Deletion/duplication and sequencing analysis is not offered for exons 6-7. **ATP8B1:** Sequencing analysis for exons 19 includes only cds +/- 10 bp. **FANCD2:** Deletion/duplication analysis is not offered for exons 14-17, 22 and sequencing analysis is not offered for exons 15-17. Sequencing analysis for exons 6, 14, 18, 20, 23, 25, 34 includes only cds +/- 10 bp. **TSFM:** Sequencing analysis is not offered for exon 5. **VPS13A:** Deletion/duplication analysis is not offered for exons 2-3, 27-28. **ATM:** Sequencing analysis for exons 6, 24, 43 includes only cds +/- 10 bp. **PTPRC:** Sequencing analysis is not offered for exons 3, 15. **COL11A2:** Deletion/duplication analysis is not offered for exon 36. **DDX11:** NM_030653.3:c.1763-1G>C variant only. **ANO10:** Sequencing analysis for exons 8 includes only cds +/- 0 bp. **TBCE:** Sequencing analysis for exons 2 includes only cds +/- 10 bp. **TYR:** Deletion/duplication and sequencing analysis is not offered for exon 5. **SMN1:** Systematic exon numbering is used for all genes, including SMN1, and for this reason the exon typically referred to as exon 7 in the literature (PMID: 8838816) is referred to as exon 8 in this report. This assay unambiguously detects SMN1 exon 8 copy number. The presence of the g.27134T>G variant (also known as c.*3+80T>G) is reported if SMN1 copy number = 2. SMN1 or SMN2: NM_000344.3:c.*3+80T>G variant only. **AMN:** Deletion/duplication analysis is not offered for exon 1. **GBA:** c.84dupG (p.Leu29Alafs*18),



Patient name: 7110 Donor

DOB: [REDACTED]

Invitae #: [REDACTED]

c.115+1G>A (Splice donor), c.222_224delTAC (p.Thr75del), c.475C>T (p.Arg159Trp), c.595_596delCT (p.Leu199Aspfs*62), c.680A>G (p.Asn227Ser), c.721G>A (p.Gly241Arg), c.754T>A (p.Phe252Ile), c.1226A>G (p.Asn409Ser), c.1246G>A (p.Gly416Ser), c.1263_1317del (p.Leu422Profs*4), c.1297G>T (p.Val433Leu), c.1342G>C (p.Asp448His), c.1343A>T (p.Asp448Val), c.1448T>C (p.Leu483Pro), c.1504C>T (p.Arg502Cys), c.1505G>A (p.Arg502His), c.1603C>T (p.Arg535Cys), c.1604G>A (p.Arg535His) variants only. Rarely, sensitivity to detect these variants may be reduced. When sensitivity is reduced, zygosity may be reported as "unknown". OAT: Deletion/duplication analysis is not offered for exon 2. CFTR: Sequencing analysis for exons 7 includes only cds +/- 10 bp.

This report has been reviewed and approved by:



Mei Zhu, Ph.D., FACMG
Clinical Molecular Geneticist

7110, Donor

Patient ID:

Specimen ID:

DOB:

Age:

Sex: Male

Patient Report

Account Number:

Ordering Physician:



Date Collected: 02/09/2023

Date Received: 02/09/2023

Date Reported: 03/01/2023

Fasting: No

Ordered Items: CBC With Differential/Platelet; Chromosome, Blood, Routine; Hgb Fractionation Cascade; Venipuncture; Count 15-20 cells, 2 Karyotype; Chromosome Blood Routine 88230

General Comments & Additional Information

A courtesy copy of this report has been sent to
7865138125.

Date Collected: 02/09/2023

CBC With Differential/Platelet

| Test | Current Result and Flag | Previous Result and Date | Units | Reference Interval |
|--------------------------------------|-------------------------|--------------------------|----------|--------------------|
| WBC ⁰¹ | 4.4 | | x10E3/uL | 3.4-10.8 |
| RBC ⁰¹ | 4.87 | | x10E6/uL | 4.14-5.80 |
| Hemoglobin ⁰¹ | 14.4 | | g/dL | 13.0-17.7 |
| Hematocrit ⁰¹ | 43.3 | | % | 37.5-51.0 |
| MCV ⁰¹ | 89 | | fL | 79-97 |
| MCH ⁰¹ | 29.6 | | pg | 26.6-33.0 |
| MCHC ⁰¹ | 33.3 | | g/dL | 31.5-35.7 |
| RDW ⁰¹ | 12.4 | | % | 11.6-15.4 |
| Platelets ⁰¹ | 203 | | x10E3/uL | 150-450 |
| Neutrophils ⁰¹ | 63 | | % | Not Estab. |
| Lymphs ⁰¹ | 27 | | % | Not Estab. |
| Monocytes ⁰¹ | 8 | | % | Not Estab. |
| Eos ⁰¹ | 1 | | % | Not Estab. |
| Basos ⁰¹ | 1 | | % | Not Estab. |
| Neutrophils (Absolute) ⁰¹ | 2.8 | | x10E3/uL | 1.4-7.0 |
| Lymphs (Absolute) ⁰¹ | 1.2 | | 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 | 0.0 | | x10E3/uL | 0.0-0.2 |
| Immature Granulocytes ⁰¹ | 0 | | % | Not Estab. |
| Immature Grans (Abs) ⁰¹ | 0.0 | | x10E3/uL | 0.0-0.1 |

Chromosome, Blood, Routine

| Test | Current Result and Flag | Previous Result and Date | Units | Reference Interval |
|---|-------------------------|--------------------------|-------|--------------------|
| Specimen Type ⁰² | Comment: BLOOD | | | |
| Cells Counted ⁰² | 20 | | | |
| Cells Analyzed ⁰² | 20 | | | |
| Cells Karyotyped ⁰² | 2 | | | |
| GTG Band Resolution Achieved ⁰² | 500 | | | |
| Cytogenetic Result ⁰² | Comment: 46, XY | | | |

Date Created and Stored 03/01/23 1521 ET **Final Report** Page 1 of 3

7110, Donor

Patient ID:

Specimen ID:

DOB:

Age:

Sex: Male

Patient Report

Account Number:

Ordering Physician:



Date Collected: 02/09/2023

Chromosome, Blood, Routine (Cont.)Interpretation⁰²

Comment:

NORMAL MALE KARYOTYPE

Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTG banding pattern in all cells observed.

This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.

Technical Component-Processing performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr, Research Triangle Park, NC 27709. Medical Director, Anjen Chenn, M.D., Ph.D.

Technical Component-Chromosome analysis performed by LabCorp, CLIA 45D0674994. 7207 North Gessner Rd., Houston, TX 77040. Laboratory Director, Venkateswara R Potluri PhD.

Director Review:⁰²

Comment:

PATRICK A. LENNON, PHD, FACMG

PDF

Hgb Fractionation Cascade

| Test | Current Result and Flag | Previous Result and Date | Units | Reference Interval |
|--|-------------------------|--------------------------|-------|--------------------|
| Hgb Fractionation by CE: ⁰¹ | | | | |
| Hgb F ⁰¹ | 0.0 | | % | 0.0-2.0 |
| Hgb A ⁰¹ | 97.6 | | % | 96.4-98.8 |
| Hgb A2 ⁰¹ | 2.4 | | % | 1.8-3.2 |
| Hgb S ⁰¹ | 0.0 | | % | 0.0 |

Interpretation:⁰¹

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).

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: TA - Labcorp Tampa 5610 W LaSalle Street, Tampa, FL, 33607-1770 Dir: Sean Farrier, MD

02: YU - Labcorp RTP 1904 TW Alexander Drive Ste C, RTP, NC, 27709-0153 Dir: Anjen Chenn, MD PhD

For Inquiries, the physician may contact Branch: 713-856-8288 Lab: 800-877-5227

Date Created and Stored 03/01/23 1521 ET **Final Report** Page 2 of 3



Patient Information:

7110, Donor

DOB: [REDACTED]

Sex: M

MR#: 7110

Patient#: [REDACTED]

Accession:

[REDACTED]

Specimen Type: DNA

Collected: Not provided

Received Date: Jan 23, 2025

Authorized Date: Jan 26, 2025

Physician:

Seitz, Suzanne

ATTN: Seitz, Suzanne

Fairfax Cryobank

3015 Williams Drive

Fairfax, VA 22031

Phone:

Fax:

Laboratory:

Fulgent Therapeutics LLC

CAP#: 8042697

CLIA#: 05D2043189

Laboratory Director:

Dr. Amar Jariwala

Report Date: **Feb 10, 2025**

Final Report

TEST PERFORMED

ABCC6 Single Gene

(1 Gene Panel: *ABCC6*; gene sequencing with deletion and duplication analysis)

RESULTS:

No clinically significant sequence or copy-number variants 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 of the sort not queried by this test or in areas not reliably assessed by this test.

INTERPRETATION:

Notes and Recommendations:

- As requested, this report only includes variants classified as Pathogenic, Likely Pathogenic, or Risk Allele at the time of analysis. If detected, this report does not include variants classified as of uncertain significance.
- Gene specific notes and limitations may be present. See below.
- These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; <https://www.nsgc.org>)
- Guide to Interpreting Genomic Reports: A Genomics Toolkit (CSER Consortium; February 2017) (<https://www.genome.gov/For-Health-Professionals/Provider-Genomics-Education-Resources#hep>)

GENES TESTED:

ABCC6 Single Gene

1 genes tested (100.00% at >20x).

ABCC6

Gene Specific Notes and Limitations

ABCC6: Significant pseudogene interference in exons 1-9 of the *ABCC6* gene has been known to occur and may interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses.

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 identified by NGS are confirmed by an orthogonal method (qPCR or MLPA), unless exceeding an internally specified and validated quality score, beyond which deletions and duplications are considered real without further confirmation. New York patients: diagnostic findings are confirmed by Sanger, MLPA, or qPCR; exception SNV variants in genes for which confirmation of NGS results has been performed ≥ 10 times may not be confirmed if identified with high quality by NGS. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

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 this individual's phenotype, and negative results do not rule out a genetic cause for the indication for testing. 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 designed and validated for detection of germline variants only. It 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 (eg. trinucleotide or hexanucleotide repeat expansion). 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 for copy number variants, 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 are two or more contiguous exons in size; 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.

SIGNATURE:

A handwritten signature in black ink, appearing to read 'Zhenbin Chen'.

Zhenbin Chen, Ph.D., CGMB, FACMG on 2/10/2025
Laboratory Director, Fulgent



DISCLAIMER:

This test was developed and its performance characteristics determined by **Fulgent Therapeutics LLC**. 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.



Patient Information:

7110, Donor

DOB: [REDACTED]

Sex: M

Accession:

[REDACTED]

Specimen Type: DNA

Collected: Not Provided

Partner Information:

Not Tested

Accession:

N/A

Physician:

Wieloch, Shannon

GC: Wieloch, Shannon

Fairfax Cryobank

3015 Williams Drive #110

Fairfax, VA 22031

Phone: [REDACTED]

Laboratory:

Fulgent Therapeutics LLC

CAP#: 8042697

CLIA#: 05D2043189

Laboratory Director:

Dr. Amar Jariwala

Report Date: Jun 17, 2025

FINAL RESULTS



No carrier mutations identified

TEST PERFORMED

Single Gene Carrier Screening: SH3TC2

(1 Gene Panel: SH3TC2; 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 Carrier Screening Panel - Gene

This analysis was run using the Custom Beacon 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.

SH3TC2

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 (\pm 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

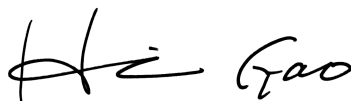
These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of 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.



Gene Specific Notes and Limitations

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

SIGNATURE:

A handwritten signature in black ink that reads "H. Gao".

Dr. Harry Gao, DABMG, FACMG on 06/17/2025
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 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.

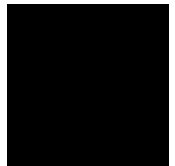
4399 Santa Anita Ave.
El Monte, CA, 91731
(p) 626-350-0537 (f) 626-454-1667
info@fulgentgenetics.com
www.fulgentgenetics.com



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:

7110, Donor

DOB: [REDACTED]

Sex: M

MR#: 7110

Patient#: F [REDACTED]

Accession:

[REDACTED]
Specimen Type: DNA
Collected: Not provided
Received Date: Jan 23, 2025
Authorized Date: Jun 17, 2025

Physician:

Wieloch, Shannon
GC: Wieloch, Shannon
Fairfax Cryobank
3015 Williams Drive #110
Fairfax, VA 22031
Phone: 7038763869
Fax:

Laboratory:

Fulgent Therapeutics LLC
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Amar Jariwala
Report Date: **Jun 30, 2025**

Final Report

TEST PERFORMED

Custom NGS Panel - 2 Genes

(2 Gene Panel: *MFN2* and *VWF*; gene sequencing with deletion and duplication analysis)

RESULTS:

No clinically significant sequence or copy-number variants 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 of the sort not queried by this test or in areas not reliably assessed by this test.

INTERPRETATION:

Notes and Recommendations:

- As requested, this report only includes variants classified as Pathogenic, Likely Pathogenic, or Risk Allele at the time of analysis. If detected, this report does not include variants classified as of uncertain significance.
- Gene specific notes and limitations may be present. See below.
- These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; <https://www.nsgc.org>)
- Guide to Interpreting Genomic Reports: A Genomics Toolkit (CSER Consortium; February 2017) (<https://www.genome.gov/For-Health-Professionals/Provider-Genomics-Education-Resources#hep>)

GENES TESTED:

Custom NGS Panel - 2 Genes

2 genes tested (98.52% at >20x).

MFN2, *VWF*

Gene Specific Notes and Limitations

VWF: Significant pseudogene interference in exons 23-34 of the *VWF* gene has been known to occur and may interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses.



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, 98.54% and 98.52% 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 identified by NGS are confirmed by an orthogonal method (qPCR or MLPA), unless exceeding an internally specified and validated quality score, beyond which deletions and duplications are considered real without further confirmation. Bioinformatics: The FPLMv2.0 pipeline was used to analyze this specimen.

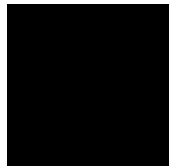
LIMITATIONS:

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 this individual's phenotype, and negative results do not rule out a genetic cause for the indication for testing. 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 designed and validated for detection of germline variants only. It 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 (eg. trinucleotide or hexanucleotide repeat expansion). 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 for copy number variants, 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 are two or more contiguous exons in size; 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.

SIGNATURE:



Geetu Mendiratta-Vij, PhD, FACMG, CGMBS on 06/30/2025
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 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.