



Donor 7276

Genetic Testing Summary

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

Last Updated: 02/11/2026

Donor Reported Ancestry: Danish, German, Welsh

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- 549 diseases by gene sequencing.	Carrier: Glycogen Storage Disease, Type 2 (Pompe Disease) (GAA) Negative for other genes sequenced.	Partner testing is recommended before using this donor.
Special Testing		
Gene: GNRHR	No carrier mutations identified via gene 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 Information

Patient Name: Donor 7276

Date Of Birth: [REDACTED]

Gender: Male

Ethnicity: Northern European
Caucasian

Patient ID: [REDACTED]

Medical Record #: 7276-[REDACTED]

Collection Kit: [REDACTED]

Accession ID: N/A

Case File ID: [REDACTED]

Test Information

Ordering Physician: [REDACTED]

Clinic Information: Fairfax Cryobank

Phone: N/A

Report Date: 06/27/2024

Sample Collected: 06/13/2024

Sample Received: 06/14/2024

Sample Type: Blood

**CARRIER SCREENING REPORT**

ABOUT THIS SCREEN: Horizon™ is a carrier screen for specific autosomal recessive and X-linked diseases. This information can help patients learn their risk of having a child with specific genetic conditions.

ORDER SELECTED: The Horizon Custom panel was ordered for this patient. Males are not screened for X-linked diseases

FINAL RESULTS SUMMARY:**CARRIER for Glycogen Storage Disease, Type 2 (Pompe Disease)**

Positive for the pathogenic variant c.-32-13T>G in the GAA gene. Please note, individuals with this GAA gene variant tend to have a later onset of the disease, slower progression and longer lifespan. While the age of onset can vary between infancy to more than 50 years of age, no patients with this variant have been reported to develop the classic infantile form of Pompe disease. Homozygotes for this variant are not expected to be affected with infantile Pompe disease, unless this variant is present as part of a complex allele. If this individual's partner is a carrier for GLYCOGEN STORAGE DISEASE, TYPE 2 (POMPE DISEASE), their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

Negative for 548 out of 549 diseases

No other pathogenic variants were detected in the genes that were screened. The patient's remaining carrier risk after the negative screening results is listed for each disease/gene on the Horizon website at <https://www.natera.com/panel-option/h-all/>. Please see the following pages of this report for a comprehensive list of all conditions included on this individual's screen.

Carrier screening is not diagnostic and may not detect all possible pathogenic variants in a given gene.

RECOMMENDATIONS

Individuals who would like to review their Horizon report with a Natera Laboratory Genetic Counselor may schedule a telephone genetic information session by calling 650-249-9090 or visiting naterasession.com. Clinicians with questions may contact Natera at 650-249-9090 or email support@natera.com. Individuals with positive results may wish to discuss these results with family members to allow them the option to be screened. Comprehensive genetic counseling to discuss the implications of these test results and possible associated reproductive risk is recommended.

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Patient Information

Patient Name: Donor 7276

Test Information

Ordering Physician: [REDACTED]



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GLYCOGEN STORAGE DISEASE, TYPE 2 (POMPE DISEASE)**Understanding Your Horizon Carrier Screen Results****What is Glycogen Storage Disease, Type 2 (Pompe Disease)?**

Glycogen Storage Disease, Type 2 (GSD2), also known as Pompe Disease, is an inherited disorder that causes progressive weakness in the muscles used for movement and breathing. People with GSD2 are missing an enzyme that breaks down glycogen, a stored form of sugar used for energy by the muscles. As a result, glycogen builds up in the body, mostly in the muscles, and damages these cells. There are three main forms of GSD2: classic infantile-onset, non-classic infantile-onset, and late-onset. The classic infantile form is the most common and most severe type of GSD2. Infants may appear normal at birth but begin to show symptoms in the first few months of life. Symptoms include decreased muscle tone (hypotonia), muscle weakness, difficulty feeding, delayed growth, breathing problems, enlarged liver and heart, and sometimes an enlarged tongue. GSD2 progresses quickly and most untreated infants will die within the first year of life. Enzyme replacement therapy may slow down the progression of heart disease and muscle weakness. The non-classic infantile form of GSD2 has symptoms that begin around the age of one and include delayed development, muscle weakness, enlarged heart, and breathing problems. Without treatment, children with this form usually die in early childhood. Symptoms of late-onset GSD2 can begin at any time from childhood to adulthood. Symptoms include progressive muscle weakness and problems with breathing, often leading to the need for wheelchair and breathing machine assistance. This form of the disease progresses more slowly, especially with enzyme-replacement therapy. Clinical trials involving potential new treatments for either form of this condition may be available (see www.clinicaltrials.gov).

What causes Glycogen Storage Disease, Type 2 (Pompe Disease)?

GSD2 is caused by gene changes, or mutations, in both copies of the GAA gene pair. These mutations cause the genes to not work properly or not work at all. When both copies of this gene do not work correctly, it leads to the symptoms described above. GSD2 is inherited in an autosomal recessive manner. This means that, in most cases, both parents must be carriers of a mutation in one copy of the GAA gene to have a child with GSD2. People who are carriers for GSD2 disease are usually healthy and do not have symptoms nor do they have GSD2 themselves. Usually a child inherits two copies of each gene, one copy from the mother and one copy from the father. If the mother and father are both carriers for GSD2 there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their GAA gene mutations to the child, who will then have GSD2. Individuals found to carry more than one mutation for GSD2 should discuss their risk for having an affected child and any potential effects on their own health with their health care provider. There are many other forms of Glycogen Storage Disease, each caused by mutations in different genes. People who are carriers for GSD2 are not likely to be at increased risk for having a child with these other forms.

What can I do next?

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website (www.nsgc.org). Your siblings and other relatives are at increased risk to also have this mutation. You are encouraged to inform your family members of your test results as they may wish to consider being tested themselves. If you are pregnant, your partner can have carrier screening for GSD2 ordered by a health care professional. If your partner is not found to be a carrier for GSD2 your risk of having a child with GSD2 is greatly reduced. Couples at risk of having a baby with GSD2 can opt to have prenatal diagnosis done through chorionic villus sampling (CVS) or amniocentesis during pregnancy or can choose to have the baby tested after birth for GSD2. If you are not yet pregnant, your partner can have carrier screening for GSD2 ordered by a health care professional. If your partner is found to be a carrier for GSD2, you have several reproductive options to consider:

- Natural pregnancy with or without prenatal diagnosis of the fetus or testing the baby after birth for GSD2
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for GSD2
- Adoption or use of a sperm or egg donor who is not a carrier for GSD2

What resources are available?

- Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/pompe-disease>
- Association for Glycogen Storage Disease: www.agsdus.org
- Prenatal diagnosis done through CVS: <http://www.marchofdimes.org/chorionic-villus-sampling.aspx>
- Prenatal diagnosis done through Amniocentesis: <http://www.marchofdimes.org/amniocentesis.aspx>
- PGD with IVF: <http://www.natera.com/spectrum>

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**VARIANT DETAILS****GAA, c.-32-13T>G, heterozygous, pathogenic**

- The c.-32-13T>G variant in the GAA gene has been observed at a frequency of 0.3401% in the gnomAD v2.1.1 dataset.
- This variant has been reported in a homozygous state or in conjunction with another variant in individual(s) with late-onset glycogen storage disease, type II (PMID: 21439876, 24844452).
- This variant has been reported in ClinVar [ID: 4027].

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DISEASES SCREENED

Below is a list of all diseases screened and the result. Certain conditions have unique patient-specific numerical values, therefore, results for those conditions are formatted differently.

Autosomal Recessive

- 1**
17-BETA HYDROXYSTEROID DEHYDROGENASE 3 DEFICIENCY (*HSD17B3*) **negative**
- 3**
3-BETA-HYDROXYSTEROID DEHYDROGENASE TYPE II DEFICIENCY (*HSD3B2*) **negative**
3-HYDROXY-3-METHYLGLUTARYL-COENZYME A LYASE DEFICIENCY (*HMGL*) **negative**
3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (*HADH*) **negative**
3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY (*MCCC2*) **negative**
3-PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY (*PHGDH*) **negative**

- 5**
5-ALPHA-REDUCTASE DEFICIENCY (*SRD5A2*) **negative**

- 6**
6-PYRUVYL-TETRAHYDROPTERIN SYNTHASE (PTPS) DEFICIENCY (*PTS*) **negative**

- A**
ABCA4-RELATED CONDITIONS (*ABCA4*) **negative**
ABETALIPOPROTEINEMIA (*MTPP*) **negative**
ACHONDROGENESIS, TYPE 1B (*SLC26A2*) **negative**
ACHROMATOPSIA, CNGB3-RELATED (*CNGB3*) **negative**
ACRODERMATITIS ENTEROPATHICA (*SLC39A4*) **negative**
ACTION MYOCLONUS-RENAL FAILURE (AMRF) SYNDROME (*SCARB2*) **negative**
ACUTE INFANTILE LIVER FAILURE, TRMU-RELATED (*TRMU*) **negative**
ACYL-COA OXIDASE I DEFICIENCY (*ACOX1*) **negative**
AICARDI-GOUTIERES SYNDROME (*SAMHD1*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2A-RELATED (*RNASEH2A*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2B-RELATED (*RNASEH2B*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2C-RELATED (*RNASEH2C*) **negative**
AICARDI-GOUTIERES SYNDROME, TREX1-RELATED (*TREX1*) **negative**
ALPHA-MANNOSIDOSIS (*MAN2B1*) **negative**
ALPHA-THALASSEMIA (*HBA1/HBA2*) **negative**
ALPORT SYNDROME, COL4A3-RELATED (*COL4A3*) **negative**
ALPORT SYNDROME, COL4A4-RELATED (*COL4A4*) **negative**
ALSTROM SYNDROME (*ALMS1*) **negative**
AMISH INFANTILE EPILEPSY SYNDROME (*ST3GAL5*) **negative**
ANDERMANN SYNDROME (*SLC12A6*) **negative**
ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (*GATM*) **negative**
ARGININEMIA (*ARG1*) **negative**
ARGININOSUCCINATE LYASE DEFICIENCY (*ASL*) **negative**
AROMATASE DEFICIENCY (*CYP19A1*) **negative**
ASPARAGINE SYNTHETASE DEFICIENCY (*ASNS*) **negative**
ASPARTYLGLYCOSAMINURIA (*AGA*) **negative**
ATAXIA WITH VITAMIN E DEFICIENCY (*TPA*) **negative**
ATAXIA-TELANGIECTASIA (*ATM*) **negative**
ATAXIA-TELANGIECTASIA-LIKE DISORDER 1 (*MRE11*) **negative**
ATRANSFERRINEMIA (*TF*) **negative**
AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (*SLC35A3*) **negative**
AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (*AIRE*) **negative**
AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (*ARCI*), SLC27A4-RELATED (*SLC27A4*) **negative**
AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (*SACS*) **negative**

- B**
BARDET-BIEDL SYNDROME, ARL6-RELATED (*ARL6*) **negative**
BARDET-BIEDL SYNDROME, BBS10-RELATED (*BBS10*) **negative**
BARDET-BIEDL SYNDROME, BBS12-RELATED (*BBS12*) **negative**
BARDET-BIEDL SYNDROME, BBS1-RELATED (*BBS1*) **negative**
BARDET-BIEDL SYNDROME, BBS2-RELATED (*BBS2*) **negative**
BARDET-BIEDL SYNDROME, BBS4-RELATED (*BBS4*) **negative**
BARDET-BIEDL SYNDROME, BBS5-RELATED (*BBS5*) **negative**
BARDET-BIEDL SYNDROME, BBS7-RELATED (*BBS7*) **negative**
BARDET-BIEDL SYNDROME, BBS9-RELATED (*BBS9*) **negative**
BARDET-BIEDL SYNDROME, TTC8-RELATED (*TTC8*) **negative**
BARE LYMPHOCYTE SYNDROME, CIITA-RELATED (*CIITA*) **negative**
BARTTER SYNDROME, BSND-RELATED (*BSND*) **negative**
BARTTER SYNDROME, KCNJ1-RELATED (*KCNJ1*) **negative**
BARTTER SYNDROME, SLC12A1-RELATED (*SLC12A1*) **negative**
BATTEN DISEASE, CLN3-RELATED (*CLN3*) **negative**
BETA-HEMOGLOBINOPATHIES (*HBB*) **negative**
BETA-KETOTHIOLENE DEFICIENCY (*ACAT1*) **negative**
BETA-MANNOSIDOSIS (*MANBA*) **negative**
BETA-UREIDOPROPIONASE DEFICIENCY (*UPB1*) **negative**
BILATERAL FRONTOPARIETAL POLYMICROGYRIA (*GPR56*) **negative**

- BIOTINIDASE DEFICIENCY (*BTD*) **negative**
BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBGD) (*SLC19A3*) **negative**
BLOOM SYNDROME (*BLM*) **negative**
BRITTLE CORNEA SYNDROME 1 (*ZNF469*) **negative**
BRITTLE CORNEA SYNDROME 2 (*PRDM5*) **negative**

- C**
CANAVAN DISEASE (*ASPA*) **negative**
CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (*CPS1*) **negative**
CARNITINE DEFICIENCY (*SLC22A5*) **negative**
CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY (*CPT1A*) **negative**
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (*CPT2*) **negative**
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (*SLC25A20*) **negative**
CARPENTER SYNDROME (*RAB23*) **negative**
CARTILAGE-HAIR HYPOPLASIA (*RMRP*) **negative**
CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (*CASQ2*) **negative**
CD59-MEDIATED HEMOLYTIC ANEMIA (*CD59*) **negative**
CEP152-RELATED MICROCEPHALY (*CEP152*) **negative**
CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA (CEDNIK) SYNDROME (*SNAP29*) **negative**
CEREBROTENDINOUS XANTHOMATOSIS (*CYP27A1*) **negative**
CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE C (*PLEKHG5*) **negative**
CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (*NDRG1*) **negative**
CHEDIAK-HIGASHI SYNDROME (*LYST*) **negative**
CHOREOACANTHOCYTOSIS (*VPS13A*) **negative**
CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (*CYBA*) **negative**
CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (*NCF2*) **negative**
CILIOPATHIES, RPGRIP1L-RELATED (*RPGRIP1L*) **negative**
CITRIN DEFICIENCY (*SLC25A13*) **negative**
CITRULLINEMIA, TYPE 1 (*ASS1*) **negative**
CLN10 DISEASE (*CTSD*) **negative**
COHEN SYNDROME (*VPS13B*) **negative**
COL11A2-RELATED CONDITIONS (*COL11A2*) **negative**
COMBINED MALONIC AND METHYLMALONIC ACIDURIA (*ACSF3*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (*GFM1*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (*TSFM*) **negative**
COMBINED PITUITARY HORMONE DEFICIENCY 1 (*POU1F1*) **negative**
COMBINED PITUITARY HORMONE DEFICIENCY-2 (*PROP1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (*CYP11B1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (*CYP17A1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (*CYP21A2*) **negative**
CONGENITAL ADRENAL INSUFFICIENCY, CYP11A1-RELATED (*CYP11A1*) **negative**
CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (*MPL*) **negative**
CONGENITAL CHRONIC DIARRHEA (*DGAT1*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, ALG1-RELATED (*ALG1*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-Related (*PMM2*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (*MPL*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (*ALG6*) **negative**
CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (*SEC23B*) **negative**
CONGENITAL FINNISH NEPHROSIS (*NPHS1*) **negative**
CONGENITAL HYDROCEPHALUS 1 (*CCDC88C*) **negative**
CONGENITAL HYPERINSULINISM, KCNJ11-Related (*KCNJ11*) **negative**
CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (CIPA) (*NTRK1*) **negative**
CONGENITAL MYASTHENIC SYNDROME, CHAT-RELATED (*CHAT*) **negative**
CONGENITAL MYASTHENIC SYNDROME, CHRNE-RELATED (*CHRNE*) **negative**
CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED (*COLQ*) **negative**
CONGENITAL MYASTHENIC SYNDROME, DOK7-RELATED (*DOK7*) **negative**
CONGENITAL MYASTHENIC SYNDROME, RAPSIN-RELATED (*RAPSN*) **negative**
CONGENITAL NEPHROTIC SYNDROME, PLCE1-RELATED (*PLCE1*) **negative**
CONGENITAL NEUTROPENIA, G6PC3-RELATED (*G6PC3*) **negative**
CONGENITAL NEUTROPENIA, HAX1-RELATED (*HAX1*) **negative**
CONGENITAL NEUTROPENIA, VPS45-RELATED (*VPS45*) **negative**
CONGENITAL SECRETORY CHLORIDE DIARRHEA 1 (*SLC26A3*) **negative**
CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS (*SLC4A11*) **negative**
CORTICOSTERONE METHYLOXIDASE DEFICIENCY (*CYP11B2*) **negative**
COSTEFF SYNDROME (3-METHYLGLUTACONIC ACIDURIA, TYPE 3) (*OPA3*) **negative**
CRB1-RELATED RETINAL DYSTROPHIES (*CRB1*) **negative**
CYSTIC FIBROSIS (*CFTR*) **negative**
CYSTINOSIS (*CTNS*) **negative**
CYTOCHROME C OXIDASE DEFICIENCY, PET100-RELATED (*PET100*) **negative**
CYTOCHROME P450 OXIOREDUCTASE DEFICIENCY (*POR*) **negative**

- D**
D-BIFUNCTIONAL PROTEIN DEFICIENCY (*HSD17B4*) **negative**

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D
 DEAFNESS, AUTOSOMAL RECESSIVE 77 (LOXHD1) **negative**
 DIHYDROPTERIDINE REDUCTASE (DHPR) DEFICIENCY (QDPR) **negative**
 DONNAI-BARROW SYNDROME (LRP2) **negative**
 DUBIN-JOHNSON SYNDROME (ABCC2) **negative**
 DYSKERATOSIS CONGENITA SPECTRUM DISORDERS (TERT) **negative**
 DYSKERATOSIS CONGENITA, RTEL1-RELATED (RTEL1) **negative**
 DYSTROPHIC EPIDERMOLYSIS BULLOSA, COL7A1-Related (COL7A1) **negative**

E
 EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY, CAD-RELATED (CAD) **negative**
 EHLERS-DANLOS SYNDROME TYPE VI (PLOD1) **negative**
 EHLERS-DANLOS SYNDROME, CLASSIC-LIKE, TNXB-RELATED (TNXB) **negative**
 EHLERS-DANLOS SYNDROME, TYPE VII C (ADAMT52) **negative**
 ELLIS-VAN CREVELD SYNDROME, EVC2-RELATED (EVC2) **negative**
 ELLIS-VAN CREVELD SYNDROME, EVC-RELATED (EVC) **negative**
 ENHANCED 5- CONE SYNDROME (NR2E3) **negative**
 EPIMERASE DEFICIENCY (GALACTOSEMIA TYPE III) (GALE) **negative**
 EPIPHYSEAL DYSPLASIA, MULTIPLE, 7/DESBUQUOIS DYSPLASIA 1 (CANT1) **negative**
 ERCC6-RELATED DISORDERS (ERCC6) **negative**
 ERCC8-RELATED DISORDERS (ERCC8) **negative**
 ETHYLMALONIC ENCEPHALOPATHY (ETHE1) **negative**

F
 FACTOR XI DEFICIENCY (F11) **negative**
 FAMILIAL DYSAUTONOMIA (IKBKAP) **negative**
 FAMILIAL HEMOPHAGOCYtic LYMPHOHISTIOCYTOSIS, PRF1-RELATED (PRF1) **negative**
 FAMILIAL HEMOPHAGOCYtic LYMPHOHISTIOCYTOSIS, STX11-RELATED (STX11) **negative**
 FAMILIAL HEMOPHAGOCYtic LYMPHOHISTIOCYTOSIS, STXBP2-RELATED (STXBP2) **negative**
 FAMILIAL HEMOPHAGOCYtic LYMPHOHISTIOCYTOSIS, UNC13D-RELATED (UNC13D) **negative**
 FAMILIAL HYPERCHOLESTEROLEMIA, LDLRAP1-RELATED (LDLRAP1) **negative**
 FAMILIAL HYPERCHOLESTEROLEMIA, LDLR-RELATED (LDLR) **negative**
 FAMILIAL HYPERINSULINISM, ABCC8-RELATED (ABCC8) **negative**
 FAMILIAL NEPHROGENIC DIABETES INSIPIDUS, AQP2-RELATED (AQP2) **negative**
 FANCONI ANEMIA, GROUP A (FANCA) **negative**
 FANCONI ANEMIA, GROUP C (FANCC) **negative**
 FANCONI ANEMIA, GROUP D2 (FANCD2) **negative**
 FANCONI ANEMIA, GROUP E (FANCE) **negative**
 FANCONI ANEMIA, GROUP F (FANCF) **negative**
 FANCONI ANEMIA, GROUP G (FANCG) **negative**
 FANCONI ANEMIA, GROUP I (FANCI) **negative**
 FANCONI ANEMIA, GROUP J (BRIP1) **negative**
 FANCONI ANEMIA, GROUP L (FANCL) **negative**
 FARBER LIPOGRANULOMATOSIS (ASAH1) **negative**
 FOVEAL HYPOPLASIA (SLC38A8) **negative**
 FRASER SYNDROME 3, GRIP1-RELATED (GRIP1) **negative**
 FRASER SYNDROME, FRAS1-RELATED (FRAS1) **negative**
 FRASER SYNDROME, FREM2-RELATED (FREM2) **negative**
 FRIEDREICH ATAXIA (FXN) **negative**
 FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY (FBP1) **negative**
 FUCOSIDOSIS, FUCA1-RELATED (FUCA1) **negative**
 FUMARASE DEFICIENCY (FH) **negative**

G
 GABA-TRANSAMINASE DEFICIENCY (ABAT) **negative**
 GALACTOKINASE DEFICIENCY (GALACTOSEMIA, TYPE II) (GALK1) **negative**
 GALACTOSEMIA (GALT) **negative**
 GALACTOSIALIDOSIS (CTS5A) **negative**
 GAUCHER DISEASE (GBA) **negative**
 GCH1-RELATED CONDITIONS (GCH1) **negative**
 GDF5-RELATED CONDITIONS (GDF5) **negative**
 GERODERMA OSTEODYSPLASTICA (GORAB) **negative**
 GITELMAN SYNDROME (SLC12A3) **negative**
 GLANZMANN THROMBASTHENIA (ITGB3) **negative**
 GLUTARIC ACIDEMIA, TYPE 1 (GCDH) **negative**
 GLUTARIC ACIDEMIA, TYPE 2A (ETFA) **negative**
 GLUTARIC ACIDEMIA, TYPE 2B (ETFB) **negative**
 GLUTARIC ACIDEMIA, TYPE 2C (ETFDH) **negative**
 GLUTATHIONE SYNTHETASE DEFICIENCY (GSS) **negative**
 GLYCINE ENCEPHALOPATHY, AMT-RELATED (AMT) **negative**
 GLYCINE ENCEPHALOPATHY, GLDC-RELATED (GLDC) **negative**
 GLYCOGEN STORAGE DISEASE TYPE 5 (McArdle Disease) (PYGM) **negative**
 GLYCOGEN STORAGE DISEASE TYPE IXB (PHKB) **negative**
 GLYCOGEN STORAGE DISEASE TYPE IXC (PHKG2) **negative**
 GLYCOGEN STORAGE DISEASE, TYPE 1a (G6PC) **negative**
 GLYCOGEN STORAGE DISEASE, TYPE 1b (SLC37A4) **negative**
 GLYCOGEN STORAGE DISEASE, TYPE 2 (POMPE DISEASE) (GAA) **see first page**
 GLYCOGEN STORAGE DISEASE, TYPE 3 (AGL) **negative**
 GLYCOGEN STORAGE DISEASE, TYPE 4 (GBE1) **negative**
 GLYCOGEN STORAGE DISEASE, TYPE 7 (PFKM) **negative**

GRACILE SYNDROME (BCS1L) **negative**
 GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY (GAMT) **negative**

H
 HARLEQUIN ICHTHYOSIS (ABCA12) **negative**
 HEME OXYGENASE 1 DEFICIENCY (HMOX1) **negative**
 HEMOCHROMATOSIS TYPE 2A (HFE2) **negative**
 HEMOCHROMATOSIS, TYPE 3, TFR2-Related (TFR2) **negative**
 HEPATOCEREBRAL MITOCHONDRIAL DNA DEPLETION SYNDROME, MPV17-RELATED (MPV17) **negative**
 HEREDITARY FRUCTOSE INTOLERANCE (ALDOB) **negative**
 HEREDITARY HEMOCHROMATOSIS TYPE 2B (HAMP) **negative**
 HEREDITARY SPASTIC PARAPARESIS, TYPE 49 (TECPR2) **negative**
 HEREDITARY SPASTIC PARAPLEGIA, CYP7B1-RELATED (CYP7B1) **negative**
 HERMANSKY-PUDLAK SYNDROME, AP3B1-RELATED (AP3B1) **negative**
 HERMANSKY-PUDLAK SYNDROME, BLOC1S3-RELATED (BLOC1S3) **negative**
 HERMANSKY-PUDLAK SYNDROME, BLOC1S6-RELATED (BLOC1S6) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED (HPS1) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS3-RELATED (HPS3) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS4-RELATED (HPS4) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS5-RELATED (HPS5) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS6-RELATED (HPS6) **negative**
 HOLOCARBOXYLASE SYNTHETASE DEFICIENCY (HLC5) **negative**
 HOMOCYSTINURIA AND MEGALOBlastic ANEMIA TYPE CBLG (MTR) **negative**
 HOMOCYSTINURIA DUE TO DEFICIENCY OF MTHFR (MTHFR) **negative**
 HOMOCYSTINURIA, CBS-RELATED (CBS) **negative**
 HOMOCYSTINURIA, Type cblE (MTRR) **negative**
 HYDROLETHALUS SYNDROME (HYLS1) **negative**
 HYPER-IGM IMMUNODEFICIENCY (CD40) **negative**
 HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA (HHH SYNDROME) (SLC25A15) **negative**
 HYPERPHOSPHATEMIC FAMILIAL TUMORAL CALCINOSIS, GALNT3-RELATED (GALNT3) **negative**
 HYPOMYELINATING LEUKODYSTROPHY 12 (VPS11) **negative**
 HYPOPHOSPHATASIA, ALPL-RELATED (ALPL) **negative**

I
 IMERSLUND-GRÄSBECK SYNDROME 2 (AMN) **negative**
 IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES (ICF) SYNDROME, DNMT3B-RELATED (DNMT3B) **negative**
 IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES (ICF) SYNDROME, ZBTB24-RELATED (ZBTB24) **negative**
 INCLUSION BODY MYOPATHY 2 (GNE) **negative**
 INFANTILE CEREBRAL AND CEREBELLAR ATROPHY (MED17) **negative**
 INFANTILE NEPHRONOPHTHISIS (INVS) **negative**
 INFANTILE NEUROAXONAL DYSTROPHY (PLA2G6) **negative**
 ISOLATED ECTOPIA LENTIS (ADAMTSL4) **negative**
 ISOLATED SULFITE OXIDASE DEFICIENCY (SUOX) **negative**
 ISOLATED THYROID-STIMULATING HORMONE DEFICIENCY (TSHB) **negative**
 ISOVALERIC ACIDEMIA (IVD) **negative**

J
 JOHANSON-BLIZZARD SYNDROME (UBR1) **negative**
 JOUBERT SYNDROME 2 / MECKEL SYNDROME 2 (TMEM216) **negative**
 JOUBERT SYNDROME AND RELATED DISORDERS (JSRD), TMEM67-RELATED (TMEM67) **negative**
 JOUBERT SYNDROME, AHI1-RELATED (AHI1) **negative**
 JOUBERT SYNDROME, ARL13B-RELATED (ARL13B) **negative**
 JOUBERT SYNDROME, B9D1-RELATED (B9D1) **negative**
 JOUBERT SYNDROME, B9D2-RELATED (B9D2) **negative**
 JOUBERT SYNDROME, C2CD3-RELATED/OROFACIODIGITAL SYNDROME 14 (C2CD3) **negative**
 JOUBERT SYNDROME, CC2D2A-RELATED/COACH SYNDROME (CC2D2A) **negative**
 JOUBERT SYNDROME, CEP104-RELATED (CEP104) **negative**
 JOUBERT SYNDROME, CEP120-RELATED/SHORT-RIB THORACIC DYSPLASIA 13 WITH OR WITHOUT POLYDACTYLY (CEP120) **negative**
 JOUBERT SYNDROME, CEP41-RELATED (CEP41) **negative**
 JOUBERT SYNDROME, CPLANE1-RELATED / OROFACIODIGITAL SYNDROME 6 (CPLANE1) **negative**
 JOUBERT SYNDROME, CSPP1-RELATED (CSPP1) **negative**
 JOUBERT SYNDROME, INPP5E-RELATED (INPP5E) **negative**
 JUNCTIONAL EPIDERMOLYSIS BULLOSA, COL17A1-RELATED (COL17A1) **negative**
 JUNCTIONAL EPIDERMOLYSIS BULLOSA, ITGA6-RELATED (ITGA6) **negative**
 JUNCTIONAL EPIDERMOLYSIS BULLOSA, ITGB4-RELATED (ITGB4) **negative**
 JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMB3-RELATED (LAMB3) **negative**
 JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMC2-RELATED (LAMC2) **negative**
 JUNCTIONAL EPIDERMOLYSIS BULLOSA/LARYNGOONYCHOCUTANEOUS SYNDROME, LAMA3-RELATED (LAMA3) **negative**

K
 KRABBE DISEASE (GALC) **negative**

L
 LAMELLAR ICHTHYOSIS, TYPE 1 (TGM1) **negative**

Patient Information

Patient Name: Donor 7276

Test Information

Ordering Physician: [REDACTED]



Clinic Information: Fairfax Cryobank

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: 06/27/2024

L
 LARON SYNDROME (GHR) **negative**
 LEBER CONGENITAL AMAUROSIS 2 (RPE65) **negative**
 LEBER CONGENITAL AMAUROSIS TYPE AIPL1 (AIPL1) **negative**
 LEBER CONGENITAL AMAUROSIS TYPE GUCY2D (GUCY2D) **negative**
 LEBER CONGENITAL AMAUROSIS TYPE TULP1 (TULP1) **negative**
 LEBER CONGENITAL AMAUROSIS, IQCB1-RELATED/SENIOR-LOKEN SYNDROME 5 (IQCB1) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE CEP290 (CEP290) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE LCA5 (LCA5) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE RDH12 (RDH12) **negative**
 LEIGH SYNDROME, FRENCH-CANADIAN TYPE (LRPPRC) **negative**
 LETHAL CONGENITAL CONTRACTURE SYNDROME 1 (GLE1) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER (EIF2B5) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B1-RELATED (EIF2B1) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B2-RELATED (EIF2B2) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B3-RELATED (EIF2B3) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B4-RELATED (EIF2B4) **negative**
 LIG4 SYNDROME (LIG4) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 8 (TRIM32) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2A (CAPN3) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2B (DYSF) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2C (SGCG) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2D (SGCA) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2E (SGCB) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2F (SGCD) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2I (FKRP) **negative**
 LIPOAMIDE DEHYDROGENASE DEFICIENCY (DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY) (DLD) **negative**
 LIPOID ADRENAL HYPERPLASIA (STAR) **negative**
 LIPOPROTEIN LIPASE DEFICIENCY (LPL) **negative**
 LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (HADHA) **negative**
 LRAT-RELATED CONDITIONS (LRAT) **negative**
 LUNG DISEASE, IMMUNODEFICIENCY, AND CHROMOSOME BREAKAGE SYNDROME (LICS) (NSMCE3) **negative**
 LYSINURIC PROTEIN INTOLERANCE (SLC7A7) **negative**

M
 MALONYL-COA DECARBOXYLASE DEFICIENCY (MLYCD) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 1A (BCKDHA) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 1B (BCKDHB) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 2 (DBT) **negative**
 MCKUSICK-KAUFMAN SYNDROME (MKKS) **negative**
 MECKEL SYNDROME 7/NEPHRONOPHTHISIS 3 (NPHP3) **negative**
 MECKEL-GRUBER SYNDROME, TYPE 1 (MKS1) **negative**
 MECR-RELATED NEUROLOGIC DISORDER (MECR) **negative**
 MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (ACADM) **negative**
 MEDNIK SYNDROME (AP151) **negative**
 MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS (MLC1) **negative**
 MEROSIN-DEFICIENT MUSCULAR DYSTROPHY (LAMA2) **negative**
 METABOLIC ENCEPHALOPATHY AND ARRHYTHMIAS, TANGO2-RELATED (TANGO2) **negative**
 METACHROMATIC LEUKODYSTROPHY, ARSA-RELATED (ARSA) **negative**
 METACHROMATIC LEUKODYSTROPHY, PSAP-RELATED (PSAP) **negative**
 METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CBLF (LMBRD1) **negative**
 METHYLMALONIC ACIDEMIA, MCEE-RELATED (MCEE) **negative**
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CBLC (MMACHC) **negative**
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CblD (MMADHC) **negative**
 METHYLMALONIC ACIDURIA, MMAA-RELATED (MMAA) **negative**
 METHYLMALONIC ACIDURIA, MMAB-RELATED (MMAB) **negative**
 METHYLMALONIC ACIDURIA, TYPE MUT(0) (MUT) **negative**
 MEVALONIC KINASE DEFICIENCY (MVK) **negative**
 MICROCEPHALIC OSTEODYSPLASIA PRIMORDIAL DWARFISM TYPE II (PCNT) **negative**
 MICROPTHALMIA / ANOPHTHALMIA, VSX2-RELATED (VSX2) **negative**
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, ACAD9-RELATED (ACAD9) **negative**
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFA5-RELATED (NDUFA5) **negative**
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFS6-RELATED (NDUFS6) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 1 (NDUFS4) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 10 (NDUFAF2) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 17 (NDUFAF6) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 19 (FOXRED1) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 3 (NDUFS7) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 4 (NDUFV1) **negative**
 MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 2, SCO2-RELATED (SCO2) **negative**
 MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 6 (COX15) **negative**
 MITOCHONDRIAL DNA DEPLETION SYNDROME 2 (TK2) **negative**

MITOCHONDRIAL DNA DEPLETION SYNDROME 3 (DGUOK) negative
MITOCHONDRIAL MYOPATHY AND SIDEROBLASTIC ANEMIA (MLASA1) (PUS1) negative
MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (HADHB) negative
MOLYBDENUM COFACTOR DEFICIENCY TYPE B (MOC52) negative
MOLYBDENUM COFACTOR DEFICIENCY, TYPE A (MOC51) negative
MUCOLIPIDOSIS II/III A (GNPTAB) negative
MUCOLIPIDOSIS III GAMMA (GNPTG) negative
MUCOLIPIDOSIS, TYPE IV (MCOLN1) negative
MUCOPOLYSACCHARIDOSIS, TYPE I (HURLER SYNDROME) (IDUA) negative
MUCOPOLYSACCHARIDOSIS, TYPE III A (SANFILIPPO A) (SGSH) negative
MUCOPOLYSACCHARIDOSIS, TYPE III B (SANFILIPPO B) (NAGLU) negative
MUCOPOLYSACCHARIDOSIS, TYPE III C (SANFILIPPO C) (HGSNAT) negative
MUCOPOLYSACCHARIDOSIS, TYPE III D (SANFILIPPO D) (GNS) negative
MUCOPOLYSACCHARIDOSIS, TYPE IV A (MORQUIO SYNDROME) (GALNS) negative
MUCOPOLYSACCHARIDOSIS, TYPE IV B/GM1 GANGLIOSIDOSIS (GLB1) negative
MUCOPOLYSACCHARIDOSIS, TYPE IX (HYAL1) negative
MUCOPOLYSACCHARIDOSIS, TYPE VI (MARTEAUX-LAMY) (ARSB) negative
MUCOPOLYSACCHARIDOSIS, TYPE VII (GUSB) negative
MULIBREY NANISM (TRIM37) negative
MULTIPLE PTERYGIUM SYNDROME, CHRNG-RELATED/ESCOBAR SYNDROME (CHRNG) negative
MULTIPLE SULFATASE DEFICIENCY (SUMF1) negative
MUSCLE-EYE-BRAIN DISEASE, POMGNT1-RELATED (POMGNT1) negative
MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (RYL1) negative
MUSK-RELATED CONGENITAL MYASTHENIC SYNDROME (MUSK) negative
MYONEUROGASTROINTESTINAL ENCEPHALOPATHY (MNGIE) (TYMP) negative
MYOTONIA CONGENITA (CLCN1) negative

N
N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY (NAGS) negative
NEMALINE MYOPATHY, NEB-RELATED (NEB) negative
NEPHRONOPHTHISIS 1 (NPHP1) negative
NEURONAL CEROID LIPOFUSCINOSIS, CLN5-RELATED (CLN5) negative
NEURONAL CEROID LIPOFUSCINOSIS, CLN6-RELATED (CLN6) negative
NEURONAL CEROID LIPOFUSCINOSIS, CLN8-RELATED (CLN8) negative
NEURONAL CEROID LIPOFUSCINOSIS, MFSDB-RELATED (MFSDB) negative
NEURONAL CEROID LIPOFUSCINOSIS, PPT1-RELATED (PPT1) negative
NEURONAL CEROID LIPOFUSCINOSIS, TPP1-RELATED (TPP1) negative
NGLY1-CONGENITAL DISORDER OF GLYCOSYLATION (NGLY1) negative
NIEMANN-PICK DISEASE, TYPE C1 / D (NPC1) negative
NIEMANN-PICK DISEASE, TYPE C2 (NPC2) negative
NIEMANN-PICK DISEASE, TYPES A / B (SMPD1) negative
NIJMEGEN BREAKAGE SYNDROME (NBN) negative
NON-SYNDROMIC HEARING LOSS, GJB2-RELATED (GJB2) negative
NON-SYNDROMIC HEARING LOSS, MYO15A-RELATED (MYO15A) negative
NONSYNDROMIC HEARING LOSS, OTOA-RELATED (OTOA) negative
NONSYNDROMIC HEARING LOSS, OTOF-RELATED (OTOF) negative
NONSYNDROMIC HEARING LOSS, PJKV-RELATED (PJKV) negative
NONSYNDROMIC HEARING LOSS, SYNE4-RELATED (SYNE4) negative
NONSYNDROMIC HEARING LOSS, TMC1-RELATED (TMC1) negative
NONSYNDROMIC HEARING LOSS, TMPRSS3-RELATED (TMPRSS3) negative
NONSYNDROMIC INTELLECTUAL DISABILITY (CC2D1A) negative
NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (SAM9) negative

O
OCULOCUTANEOUS ALBINISM TYPE IV (SLC45A2) negative
OCULOCUTANEOUS ALBINISM TYPE, III (TYRP1) negative
OCULOCUTANEOUS ALBINISM, OCA2-RELATED (OCA2) negative
OCULOCUTANEOUS ALBINISM, TYPES 1A AND 1B (TYR) negative
ODONTO-ONYCHO-DERMAL DYSPLASIA / SCHOPF-SCHULZ-PASSARGE SYNDROME (WNT10A) negative
OMENN SYNDROME, RAG2-RELATED (RAG2) negative
ORNITHINE AMINOTRANSFERASE DEFICIENCY (OAT) negative
OSTEOGENESIS IMPERFECTA TYPE VII (CRTAP) negative
OSTEOGENESIS IMPERFECTA TYPE VIII (P3H1) negative
OSTEOGENESIS IMPERFECTA TYPE XI (FKBP10) negative
OSTEOGENESIS IMPERFECTA TYPE XIII (BMP1) negative
OSTEOPETROSIS, INFANTILE MALIGNANT, TCIRG1-RELATED (TCIRG1) negative
OSTEOPETROSIS, OSTM1-RELATED (OSTM1) negative

P
PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION (PANK2) negative
PAPILLON LEFÈVRE SYNDROME (CTSC) negative
PARKINSON DISEASE 15 (FBX07) negative
PENDRED SYNDROME (SLC26A4) negative
PERLMAN SYNDROME (DIS3L2) negative
PGM3-CONGENITAL DISORDER OF GLYCOSYLATION (PGM3) negative
PHENYLKETONURIA (PAH) negative
PIGN-CONGENITAL DISORDER OF GLYCOSYLATION (PIGN) negative
PITUITARY HORMONE DEFICIENCY, COMBINED 3 (LHX3) negative
POLG-RELATED DISORDERS (POLG) negative

Patient Information

Patient Name: Donor 7276

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Test Information

Ordering Physician: [REDACTED]

Clinic Information: Fairfax Cryobank

Report Date:

06/27/2024



P
 POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE (*PKHD1*) **negative**
 PONTOCEREBELLAR HYPOPLASIA, EXOSC3-RELATED (*EXOSC3*) **negative**
 PONTOCEREBELLAR HYPOPLASIA, RARS2-RELATED (*RARS2*) **negative**
 PONTOCEREBELLAR HYPOPLASIA, TSEN2-RELATED (*TSEN2*) **negative**
 PONTOCEREBELLAR HYPOPLASIA, TSEN54-RELATED (*TSEN54*) **negative**
 PONTOCEREBELLAR HYPOPLASIA, TYPE 1A (*VRK1*) **negative**
 PONTOCEREBELLAR HYPOPLASIA, TYPE 2D (*SEPS5*) **negative**
 PONTOCEREBELLAR HYPOPLASIA, VPS53-RELATED (*VPS53*) **negative**
 PRIMARY CILIARY DYSKINESIA, CCDC103-RELATED (*CCDC103*) **negative**
 PRIMARY CILIARY DYSKINESIA, CCDC39-RELATED (*CCDC39*) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAH11-RELATED (*DNAH11*) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAH5-RELATED (*DNAH5*) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAI1-RELATED (*DNAI1*) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAI2-RELATED (*DNAI2*) **negative**
 PRIMARY CONGENITAL GLAUCOMA/PETERS ANOMALY (*CYP1B1*) **negative**
 PRIMARY HYPEROXALURIA, TYPE 1 (*AGXT*) **negative**
 PRIMARY HYPEROXALURIA, TYPE 2 (*GRHPR*) **negative**
 PRIMARY HYPEROXALURIA, TYPE 3 (*HOGA1*) **negative**
 PRIMARY MICROCEPHALY 1, AUTOSOMAL RECESSIVE (*MCPH1*) **negative**
 PROGRESSIVE EARLY-ONSET ENCEPHALOPATHY WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM (*TBCD*) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, ABCB4-RELATED (*ABCB4*) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 1 (PFIC1) (*ATP8B1*) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 2 (ABCB11) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 4 (PFIC4) (*TJP2*) **negative**
 PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA (*CCN6*) **negative**
 PROLIDASE DEFICIENCY (*PEPD*) **negative**
 PROPIONIC ACIDEMIA, PCCA-RELATED (*PCCA*) **negative**
 PROPIONIC ACIDEMIA, PCCB-RELATED (*PCCB*) **negative**
 PSEUDOXANTHOMA ELASTICUM (*ABCC6*) **negative**
 PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (PCD) DEFICIENCY (*PCBD1*) **negative**
 PYCNOXYSTOSIS (*CTSK*) **negative**
 PYRIDOXAL 5'-PHOSPHATE-DEPENDENT EPILEPSY (*PNPO*) **negative**
 PYRIDOXINE-DEPENDENT EPILEPSY (*ALDH7A1*) **negative**
 PYRUVATE CARBOXYLASE DEFICIENCY (*PC*) **negative**
 PYRUVATE DEHYDROGENASE DEFICIENCY, PDHB-RELATED (*PDHB*) **negative**

R
 REFSUM DISEASE, PHYH-RELATED (*PHYH*) **negative**
 RENAL TUBULAR ACIDOSIS AND DEAFNESS, ATP6V1B1-RELATED (*ATP6V1B1*) **negative**
 RENAL TUBULAR ACIDOSIS, PROXIMAL, WITH OCULAR ABNORMALITIES AND MENTAL RETARDATION (*SLC4A4*) **negative**
 RETINITIS PIGMENTOSA 25 (*EYS*) **negative**
 RETINITIS PIGMENTOSA 26 (*CERKL*) **negative**
 RETINITIS PIGMENTOSA 28 (*FAM161A*) **negative**
 RETINITIS PIGMENTOSA 36 (*PRCD*) **negative**
 RETINITIS PIGMENTOSA 59 (*DHDDS*) **negative**
 RETINITIS PIGMENTOSA 62 (*MAK*) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1 (*PEX7*) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2 (*GNPAT*) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3 (*AGPS*) **negative**
 RLBP1-RELATED RETINOPATHY (*RLBP1*) **negative**
 ROBERTS SYNDROME (*ESCO2*) **negative**
 RYR1-RELATED CONDITIONS (*RYR1*) **negative**

S
 SALLA DISEASE (*SLC17A5*) **negative**
 SANDHOFF DISEASE (*HEXB*) **negative**
 SCHIMKE IMMUNOSKELETAL DYSPLASIA (*SMARCAL1*) **negative**
 SCHINDLER DISEASE (*NAGA*) **negative**
 SEGAWA SYNDROME, TH-RELATED (*TH*) **negative**
 SENIOR-LOKEN SYNDROME 4/NEPHRONOPHTHISIS 4 (*NPHP4*) **negative**
 SEPIAPTERIN REDUCTASE DEFICIENCY (*SPR*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3D-RELATED (*CD3D*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3E-RELATED (*CD3E*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), FOXN1-RELATED (*FOXN1*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), IKBKB-RELATED (*IKBKB*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), IL7R-RELATED (*IL7R*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), JAK3-RELATED (*JAK3*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), PTPRC-RELATED (*PTPRC*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), RAG1-RELATED (*RAG1*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY, ADA-Related (*ADA*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY, TYPE ATHABASKAN (*DCLRE1C*) **negative**
 SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYL (*DYNC2H1*) **negative**
 SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED (*SBDS*) **negative**
 SIALIDOSIS (*NEU1*) **negative**
 SJÖGREN-LARSSON SYNDROME (*ALDH3A2*) **negative**
 SMITH-LEMLI-OPITZ SYNDROME (*DHCR7*) **negative**
 SPASTIC PARAPLEGIA, TYPE 15 (*ZFVVE26*) **negative**

SPASTIC TETRAPLEGIA, THIN CORPUS CALLOSUM, AND PROGRESSIVE MICROCEPHALY (SPATCCM) (*SLC1A4*) **negative**
 SPG11-RELATED CONDITIONS (*SPG11*) **negative**
 SPINAL MUSCULAR ATROPHY (*SMN1*) **negative** SMN1: Two copies; g.27134T>G: absent; the absence of the g.27134T>G variant decreases the chance to be a silent (2+0) carrier.
 SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS TYPE 1 (*IGHMBP2*) **negative**
 SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 10 (*ANO10*) **negative**
 SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12 (*WVWX*) **negative**
 SPONDYLOCOSTAL DYSOSTOSIS 1 (*DLL3*) **negative**
 SPONDYLOTHORACIC DYSOSTOSIS, MESP2-Related (*MESP2*) **negative**
 STEEL SYNDROME (*COL27A1*) **negative**
 STEROID-RESISTANT NEPHROTIC SYNDROME (*NPHS2*) **negative**
 STUVE-WIEDEMANN SYNDROME (*LIFR*) **negative**
 SURF1-RELATED CONDITIONS (*SURF1*) **negative**
 SURFACTANT DYSFUNCTION, ABCA3-RELATED (*ABCA3*) **negative**

T
 TAY-SACHS DISEASE (*HEXA*) **negative**
 TBCE-RELATED CONDITIONS (*TBCE*) **negative**
 THIAMINE-RESPONSIVE MEGALOBlastic ANEMIA SYNDROME (*SLC19A2*) **negative**
 THYROID DYSHORMONOGENESIS 1 (*SLC5A5*) **negative**
 THYROID DYSHORMONOGENESIS 2A (*TPO*) **negative**
 THYROID DYSHORMONOGENESIS 3 (*TG*) **negative**
 THYROID DYSHORMONOGENESIS 6 (*DUOX2*) **negative**
 TRANSCOBALAMIN II DEFICIENCY (*TCN2*) **negative**
 TRICHOHEPATOENTERIC SYNDROME, SKIC2-RELATED (*SKIC2*) **negative**
 TRICHOHEPATOENTERIC SYNDROME, TTC37-RELATED (*TTC37*) **negative**
 TRICHOIODYSTROPHY 1/XERODERMA PIGMENTOSUM, GROUP D (*ERCC2*) **negative**
 TRIMETHYLAMINURIA (*FMO3*) **negative**
 TRIPLE A SYNDROME (*AAA5*) **negative**
 TSHR-RELATED CONDITIONS (*TSHR*) **negative**
 TYROSINEMIA TYPE III (*HPD*) **negative**
 TYROSINEMIA, TYPE 1 (*FAH*) **negative**
 TYROSINEMIA, TYPE 2 (*TAT*) **negative**

U
 USHER SYNDROME, TYPE 1B (*MYO7A*) **negative**
 USHER SYNDROME, TYPE 1C (*USH1C*) **negative**
 USHER SYNDROME, TYPE 1D (*CDH23*) **negative**
 USHER SYNDROME, TYPE 1F (*PCDH15*) **negative**
 USHER SYNDROME, TYPE 1J/DEAFNESS, AUTOSOMAL RECESSIVE, 48 (*CIB2*) **negative**
 USHER SYNDROME, TYPE 2A (*USH2A*) **negative**
 USHER SYNDROME, TYPE 2C (*ADGRV1*) **negative**
 USHER SYNDROME, TYPE 3 (*CLRN1*) **negative**

V
 VERY LONG-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (*ACADVL*) **negative**
 VICI SYNDROME (*EPG5*) **negative**
 VITAMIN D-DEPENDENT RICKETS, TYPE 1A (*CYP27B1*) **negative**
 VITAMIN D-RESISTANT RICKETS TYPE 2A (*VDR*) **negative**
 VLDLR-ASSOCIATED CEREBELLAR HYPOPLASIA (*VLDLR*) **negative**

W
 WALKER-WARBURG SYNDROME, CRPPA-RELATED (*CRPPA*) **negative**
 WALKER-WARBURG SYNDROME, FKTN-RELATED (*FKTN*) **negative**
 WALKER-WARBURG SYNDROME, LARGE1-RELATED (*LARGE1*) **negative**
 WALKER-WARBURG SYNDROME, POMT1-RELATED (*POMT1*) **negative**
 WALKER-WARBURG SYNDROME, POMT2-RELATED (*POMT2*) **negative**
 WARSAW BREAKAGE SYNDROME (*DDX11*) **negative**
 WERNER SYNDROME (*WRN*) **negative**
 WILSON DISEASE (*ATP7B*) **negative**
 WOLCOTT-RALLISON SYNDROME (*EIF2AK3*) **negative**
 WOLMAN DISEASE (*LIPA*) **negative**
 WOODHOUSE-SAKATI SYNDROME (*DCAF17*) **negative**

X
 XERODERMA PIGMENTOSUM VARIANT TYPE (*POLH*) **negative**
 XERODERMA PIGMENTOSUM, GROUP A (*XPA*) **negative**
 XERODERMA PIGMENTOSUM, GROUP C (*XPC*) **negative**

Z
 ZELLWEGER SPECTRUM DISORDER, PEX13-RELATED (*PEX13*) **negative**
 ZELLWEGER SPECTRUM DISORDER, PEX16-RELATED (*PEX16*) **negative**
 ZELLWEGER SPECTRUM DISORDER, PEX5-RELATED (*PEX5*) **negative**
 ZELLWEGER SPECTRUM DISORDERS, PEX10-RELATED (*PEX10*) **negative**
 ZELLWEGER SPECTRUM DISORDERS, PEX12-RELATED (*PEX12*) **negative**
 ZELLWEGER SPECTRUM DISORDERS, PEX1-RELATED (*PEX1*) **negative**
 ZELLWEGER SPECTRUM DISORDERS, PEX26-RELATED (*PEX26*) **negative**
 ZELLWEGER SPECTRUM DISORDERS, PEX2-RELATED (*PEX2*) **negative**

Patient Information

Patient Name: Donor 7276

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Test Information

Ordering Physician: [REDACTED]

Clinic Information: Fairfax Cryobank

Report Date: 06/27/2024



Z
ZELLWEGER SPECTRUM DISORDERS, PEX6-RELATED (PEX6) **negative**

Patient Information
Patient Name: Donor 7276

Date Of Birth: [REDACTED]
Case File ID: [REDACTED]

Test Information
Ordering Physician: [REDACTED]
Clinic Information: Fairfax Cryobank
Report Date: 06/27/2024



Testing Methodology, Limitations, and Comments:

Next-generation sequencing (NGS)

Sequencing library prepared from genomic DNA isolated from a patient sample is enriched for targets of interest using standard hybridization capture protocols and PCR amplification (for targets specified below). NGS is then performed to achieve the standards of quality control metrics, including a minimum coverage of 99% of targeted regions at 20X sequencing depth. Sequencing data is aligned to human reference sequence, followed by deduplication, metric collection and variant calling (coding region +/- 20bp). Variants are then classified according to ACMGG/AMP standards of interpretation using publicly available databases including but not limited to ENSEMBL, HGMD Pro, ClinGen, ClinVar, 1000G, ESP and gnomAD. Variants predicted to be pathogenic or likely pathogenic for the specified diseases are reported. It should be noted that the data interpretation is based on our current understanding of the genes and variants at the time of reporting. Putative positive sequencing variants that do not meet internal quality standards or are within highly homologous regions are confirmed by Sanger sequencing or gene-specific long-range PCR as needed prior to reporting.

Copy Number Variant (CNV) analysis is limited to deletions involving two or more exons for all genes on the panel, in addition to specific known recurrent single-exon deletions. CNVs of small size may have reduced detection rate. This method does not detect gene inversions, single-exonic and sub-exonic deletions (unless otherwise specified), and duplications of all sizes (unless otherwise specified). Additionally, this method does not define the exact breakpoints of detected CNV events. Confirmation testing for copy number variation is performed by specific PCR, Multiplex Ligation-dependent Probe Amplification (MLPA), next generation sequencing, or other methodology.

This test may not detect certain variants due to local sequence characteristics, high/low genomic complexity, homologous sequence, or allele dropout (PCR-based assays). Variants within noncoding regions (promoter, 5'UTR, 3'UTR, deep intronic regions, unless otherwise specified), small deletions or insertions larger than 25bp, low-level mosaic variants, structural variants such as inversions, and/or balanced translocations may not be detected with this technology.

SPECIAL NOTES

For ABCC6, variants in exons 1-9 are not detected due to the presence of regions of high homology.

For CFTR, when the CFTR R117H variant is detected, reflex analysis of the polythymidine variations (5T, 7T and 9T) at the intron 9 branch/acceptor site of the CFTR gene will be performed.

For CYP21A2, targets were enriched using long-range PCR amplification, followed by next generation sequencing. Duplication analysis will only be performed and reported when c.955C>T (p.Q319*) is detected. Sequencing and CNV analysis may have reduced sensitivity, if variants result from complex rearrangements, in trans with a gene deletion, or CYP21A2 gene duplication on one chromosome and deletion on the other chromosome. This analysis cannot detect sequencing variants located on the CYP21A2 duplicated copy.

For DDX11, only NM_030653.3:c.1763 - 1G > C variant will be analyzed and reported.

For HBA1/HBA2, CNV analysis is offered to detect common deletions of -alpha3.7, -alpha4.2, --MED, --SEA, --FIL, --THAI, --alpha20.5, and/or HS-40.

For OTOA, variants in exons 20 - 28 are not analyzed due to high sequence homology.

For RPGRIP1L, variants in exon 23 are not detected due to assay limitation.

For SAMD9, only p.K1495E variant will be analyzed and reported.

Friedreich Ataxia (FXN)

The GAA repeat region of the FXN gene is assessed by trinucleotide PCR assay and capillary electrophoresis. Variances of +/-1 repeat for normal alleles and up to +/-3 repeats for premutation alleles may occur. For fully penetrant expanded alleles, the precise repeat size cannot be determined, therefore the approximate allele size is reported. Sequencing and copy number variants are analyzed by next-generation sequencing analysis.

Friedreich Ataxia Repeat Categories

Categories	GAA Repeat Sizes
Normal	<34
Premutation	34 - 65
Full	>65

Patient Information

Patient Name: Donor 7276

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Test Information

Ordering Physician: [REDACTED]

Clinic Information: Fairfax Cryobank

Report Date: 06/27/2024

**Spinal Muscular Atrophy (SMN1)**

The total combined copy number of SMN1 and SMN2 exon 7 is quantified based on NGS read depth. The ratio of SMN1 to SMN2 is calculated based on the read depth of a single nucleotide that distinguishes these two genes in exon 7. In addition to copy number analysis, testing for the presence or absence of a single nucleotide polymorphism (g.27134T>G in intron 7 of SMN1) associated with the presence of a SMN1 duplication allele is performed using NGS.

Ethnicity	Two SMN1 copies carrier risk before g.27134T>G testing	Carrier risk after g.27134T>G testing	
		g.27134T>G ABSENT	g.27134T>G PRESENT
Caucasian	1 in 632	1 in 769	1 in 29
Ashkenazi Jewish	1 in 350	1 in 580	LIKELY CARRIER
Asian	1 in 628	1 in 702	LIKELY CARRIER
African-American	1 in 121	1 in 396	1 in 34
Hispanic	1 in 1061	1 in 1762	1 in 140

Variant Classification

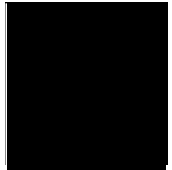
Only pathogenic or likely pathogenic variants are reported. Other variants including benign variants, likely benign variants, variants of uncertain significance, or inconclusive variants identified during this analysis may be reported in certain circumstances. Our laboratory's variant classification criteria are based on the ACMG and internal guidelines and our current understanding of the specific genes. This interpretation may change over time as more information about a gene and/or variant becomes available. Natera and its lab partner(s) may reclassify variants at certain intervals but may not release updated reports without a specific request made to Natera by the ordering provider. Natera may disclose incidental findings if deemed clinically pertinent to the test performed.

Negative Results

A negative carrier screening result reduces the risk for a patient to be a carrier of a specific disease but does not completely rule out carrier status. Please visit <https://www.natera.com/panel-option/h-all/> for a table of carrier rates, detection rates, residual risks and promised variants/exons per gene. Carrier rates before and after testing vary by ethnicity and assume a negative family history for each disease screened and the absence of clinical symptoms in the patient. Any patient with a family history for a specific genetic disease will have a higher carrier risk prior to testing and, if the disease-causing mutation in their family is not included on the test, their carrier risk would remain unchanged. Genetic counseling is recommended for patients with a family history of genetic disease so that risk figures based on actual family history can be determined and discussed along with potential implications for reproduction. Horizon carrier screening has been developed to identify the reproductive risks for monogenic inherited conditions. Even when one or both members of a couple screen negative for pathogenic variants in a specific gene, the disease risk for their offspring is not zero. There is still a low risk for the condition in their offspring due to a number of different mechanisms that are not detected by Horizon including, but not limited to, pathogenic variant(s) in the tested gene or in a different gene not included on Horizon, pathogenic variant(s) in an upstream regulator, uniparental disomy, de novo mutation(s), or digenic or polygenic inheritance.

Additional Comments

These analyses generally provide highly accurate information regarding the patient's carrier status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.



Patient Information:

7276, Donor

DOB: [REDACTED]

Sex: M

MR#: 7276

Patient#: [REDACTED]

Partner Information:

Not Tested

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: **Feb 09,2026**

Accession:

[REDACTED]

Specimen Type: DNA

Collected: Not Provided

Accession:

N/A

FINAL RESULTS



No carrier mutations identified

TEST PERFORMED

Single Gene Carrier Screening: **GNRHR**

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

INTERPRETATION:

Notes and Recommendations:

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



GENES TESTED:

Custom Beacon Carrier Screening Panel - 1 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.

GNRHR

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 100.00% and 100.00% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The FPLMv2.0 pipeline was used to analyze this specimen.

LIMITATIONS:

General Limitations

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



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 "Harry Gao".

Dr. Harry Gao, DABMG, FACMG on 02/09/2026
Laboratory Director, Fulgent

DISCLAIMER:

This test was developed, performed, and its performance characteristics determined by **Fulgent Therapeutics LLC** (CAP# 8042697, CLIA# 05D2043189), 4399 Santa Anita Ave., El Monte, CA 91731. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at **(626) 350-0537** or info@fulgentgenetics.com. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.

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

[Beacon Expanded Carrier Screening Supplemental Table](#)

