

GENETIC TESTING SUMMARY

Sperm Donor # 7820

This Genetic Testing Summary provides general information regarding a donor's genetic test results. It is not comprehensive and does not replace a clinical genetic counseling consultation or constitute medical advice. Clients should review the donor's complete genetic test results with their medical provider or fertility specialist.

Genetic testing technology and scientific information change over time. Testing is not available for every genetic condition, and Fairfax does not test for every condition for which testing is available. A negative result reduces, but does not eliminate, the risk of having an affected child.

Last Updated: 07/17/2026

Genetic Test*	Result	Comments
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Expanded Genetic Carrier Screening: 549 diseases by gene sequencing and del/dup analysis.	Carrier: Factor XI Deficiency (F11) Negative for other genes tested.	Egg source carrier screening is indicated before using this donor Carriers of Factor XI Deficiency may exhibit symptoms of this disease. Please see results for further information. Genetic counseling can be considered.

Patient Information

Patient Name: Donor 7820

Date Of Birth: [REDACTED]

Gender: Male

Ethnicity: Other

Patient ID: N/A

Medical Record #: 7820-[REDACTED]

Collection Kit: [REDACTED]

Accession ID: N/A

Case File ID: [REDACTED]

Test Information

Ordering Physician: [REDACTED]

Clinic Information: Fairfax Cryobank

Phone: [REDACTED]

Report Date: 07/29/2025

Sample Collected: 07/15/2025

Sample Received: 07/16/2025

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 Factor Xi Deficiency**

Positive for the likely pathogenic variant c.1627G>T (p.E543*) in the F11 gene. FACTOR XI DEFICIENCY carriers may exhibit mild to severe symptoms of this disease. Comprehensive genetic counseling and additional medical workup as clinically indicated should be considered. If this individual's partner is a carrier for FACTOR XI DEFICIENCY, their chance to have a child with this condition may be as high as 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](https://www.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 Name: Donor 7820

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FACTOR XI DEFICIENCY

Understanding Your Horizon Carrier Screen Results

What is Factor XI Deficiency?

Factor XI Deficiency, also called Hemophilia C, is an inherited bleeding disorder that causes mild to heavy bleeding, especially following dental tooth extraction, surgery, or trauma. Some people with Factor XI Deficiency also experience frequent nosebleeds and bruising. Women may have heavy menstrual cycles or postpartum bleeding. Most people with this condition will have mild symptoms; however, symptoms may be more severe in certain cases. Treatment with medications that help blood clot more quickly may be helpful for people with severe bleeding problems. Factor XI deficiency is considered less severe than other forms of Hemophilia (types A and B). Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Factor XI Deficiency?

Factor XI Deficiency is caused by a gene change, or mutation, in both copies of the F11 gene pair. These mutations cause the genes to not work properly or not work at all. Normal function of the F11 genes is important for making a protein that helps in blood clotting. When both copies of the F11 gene do not work correctly, it leads to the symptoms described above. When one copy of this gene works correctly and the other does not, it may cause mild bleeding problems or no symptoms at all. Factor XI Deficiency 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 F11 gene to have a child with Factor XI Deficiency. People who are carriers for Factor XI Deficiency may have some symptoms of Factor XI Deficiency such as prolonged bleeding after surgery, trauma, or tooth extraction, or they may have no symptoms at all. 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 Factor XI Deficiency, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their F11 gene mutations to the child, who will then have Factor XI Deficiency. There is a 1 in 2, or 50%, chance in each pregnancy for the child to inherit one mutation and be a carrier for Factor XI Deficiency with a chance to have mild bleeding problems. And, there is a 1 in 4, or 25% chance in each pregnancy for the child to inherit no F11 gene mutations. Individuals found to carry more than one mutation for Factor XI Deficiency should discuss their risk for having an affected child, and any risks to their own health, with their health care provider.

What can I do next?

We encourage you to share these results with your health care providers and dentist, especially if you think you may have bleeding problems. 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 Factor XI Deficiency ordered by a health care professional. If your partner is not found to be a carrier for Factor XI Deficiency, your risk of having an affected child is greatly reduced. Couples at risk of having a baby with Factor XI Deficiency 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 this condition. If you are not yet pregnant, your partner can have carrier screening for Factor XI Deficiency ordered by a health care professional. If your partner is found to be a carrier for Factor XI Deficiency, you have several reproductive options to consider:

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

What resources are available?

- National Hemophilia Foundation: <https://www.hemophilia.org/Bleeding-Disorders/Types-of-Bleeding-Disorders/Other-Factor-Deficiencies/Factor-XI>
- 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****F11, c.1627G>T (p.E543*), likely pathogenic**

- The c.1627G>T (p.E543*) variant in the F11 gene has been observed at a frequency of 0.0032% in the gnomAD v2.1.1 dataset.
- This premature termination variant is predicted to cause nonsense-mediated decay (NMD) in a gene where loss-of-function is a known mechanism of disease.
- This variant has not been described in ClinVar.

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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 (<i>HSD17B3</i>) negative	BIOTINIDASE DEFICIENCY (<i>BTD</i>) negative
3	3-BETA-HYDROXYSTEROID DEHYDROGENASE TYPE II DEFICIENCY (<i>HSD3B2</i>) negative	BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBGD) (<i>SLC19A3</i>) negative
	3-HYDROXY-3-METHYLGUTARYL-COENZYME A LYASE DEFICIENCY (<i>HMGCL</i>) negative	BLOOM SYNDROME (<i>BLM</i>) negative
	3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (<i>HADH</i>) negative	BRITTLE CORNEA SYNDROME 1 (<i>ZNF469</i>) negative
	3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY (<i>MCCC2</i>) negative	BRITTLE CORNEA SYNDROME 2 (<i>PRDM5</i>) negative
	3-PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY (<i>PHGDH</i>) negative	
5	5-ALPHA-REDUCTASE DEFICIENCY (<i>SRD5A2</i>) negative	C
6	6-PYRUVYL-TETRAHYDROPTERIN SYNTHASE (PTPS) DEFICIENCY (<i>PTS</i>) negative	CANAVAN DISEASE (<i>ASPA</i>) negative
A	ABCA4-RELATED CONDITIONS (<i>ABCA4</i>) negative	CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (<i>CPS1</i>) negative
	ABETALIPOPROTEINEMIA (<i>MTTP</i>) negative	CARNITINE DEFICIENCY (<i>SLC22A5</i>) negative
	ACHONDROGENESIS, TYPE 1B (<i>SLC26A2</i>) negative	CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY (<i>CPT1A</i>) negative
	ACHROMATOPSIA, CNGB3-RELATED (<i>CNGB3</i>) negative	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (<i>CPT2</i>) negative
	ACRODERMATITIS ENTEROPATHICA (<i>SLC39A4</i>) negative	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (<i>SLC25A20</i>) negative
	ACTION MYOCLONUS-RENAL FAILURE (AMRF) SYNDROME (<i>SCARB2</i>) negative	CARPENTER SYNDROME (<i>RAB23</i>) negative
	ACUTE INFANTILE LIVER FAILURE, TRMU-RELATED (<i>TRMU</i>) negative	CARTILAGE-HAIR HYPOPLASIA (<i>RMRP</i>) negative
	ACYL-COA OXIDASE I DEFICIENCY (<i>ACOX1</i>) negative	CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (<i>CASQ2</i>) negative
	AICARDI-GOUTIERES SYNDROME (<i>SAMHD1</i>) negative	CD59-MEDIATED HEMOLYTIC ANEMIA (<i>CD59</i>) negative
	AICARDI-GOUTIERES SYNDROME, RNASEH2A-RELATED (<i>RNASEH2A</i>) negative	CEP152-RELATED MICROCEPHALY (<i>CEP152</i>) negative
	AICARDI-GOUTIERES SYNDROME, RNASEH2B-RELATED (<i>RNASEH2B</i>) negative	CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA (CEDNIK) SYNDROME (<i>SNAP29</i>) negative
	AICARDI-GOUTIERES SYNDROME, RNASEH2C-RELATED (<i>RNASEH2C</i>) negative	CEREBROTENDINOUS XANTHOMATOSIS (<i>CYP27A1</i>) negative
	AICARDI-GOUTIERES SYNDROME, TREX1-RELATED (<i>TREX1</i>) negative	CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE C (<i>PLEKHG5</i>) negative
	ALPHA-MANNOSIDOSIS (<i>MAN2B1</i>) negative	CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (<i>NDRG1</i>) negative
	ALPHA-THALASSEMIA (<i>HBA1/HBA2</i>) negative	CHEDIAK-HIGASHI SYNDROME (<i>LYST</i>) negative
	ALPORT SYNDROME, COL4A3-RELATED (<i>COL4A3</i>) negative	CHOREOACANTHOCYTOSIS (<i>VPS13A</i>) negative
	ALPORT SYNDROME, COL4A4-RELATED (<i>COL4A4</i>) negative	CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (<i>CYBA</i>) negative
	ALSTROM SYNDROME (<i>ALMS1</i>) negative	CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (<i>NCF2</i>) negative
	AMISH INFANTILE EPILEPSY SYNDROME (<i>ST3GAL5</i>) negative	CILIOPATHIES, RPGRIP1L-RELATED (<i>RPGRIP1L</i>) negative
	ANDERMANN SYNDROME (<i>SLC12A6</i>) negative	CITRIN DEFICIENCY (<i>SLC25A13</i>) negative
	ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (<i>GATM</i>) negative	CITRULLINEMIA, TYPE 1 (<i>ASS1</i>) negative
	ARGININEMIA (<i>ARG1</i>) negative	CLN10 DISEASE (<i>CTSD</i>) negative
	ARGININOSUCCINATE LYASE DEFICIENCY (<i>ASL</i>) negative	COHEN SYNDROME (<i>VPS13B</i>) negative
	AROMATASE DEFICIENCY (<i>CYP19A1</i>) negative	COL11A2-RELATED CONDITIONS (<i>COL11A2</i>) negative
	ASPARAGINE SYNTHETASE DEFICIENCY (<i>ASN5</i>) negative	COMBINED MALONIC AND METHYLMALONIC ACIDURIA (<i>ACSF3</i>) negative
	ASPARTYLGLYCOSAMINURIA (<i>AGA</i>) negative	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (<i>GFM1</i>) negative
	ATAXIA WITH VITAMIN E DEFICIENCY (<i>TPA</i>) negative	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (<i>TSFM</i>) negative
	ATAXIA-TELANGIECTASIA (<i>ATM</i>) negative	COMBINED PITUITARY HORMONE DEFICIENCY 1 (<i>POU1F1</i>) negative
	ATAXIA-TELANGIECTASIA-LIKE DISORDER 1 (<i>MRE11</i>) negative	COMBINED PITUITARY HORMONE DEFICIENCY-2 (<i>PROPT</i>) negative
	ATRANSFERRINEMIA (<i>TF</i>) negative	CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (<i>CYP11B1</i>) negative
	AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (<i>SLC35A3</i>) negative	CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (<i>CYP17A1</i>) negative
	AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (<i>AIRE</i>) negative	CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (<i>CYP21A2</i>) negative
	AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (<i>ARCI</i>), <i>SLC27A4</i> -RELATED (<i>SLC27A4</i>) negative	CONGENITAL ADRENAL INSUFFICIENCY, CYP11A1-RELATED (<i>CYP11A1</i>) negative
	AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (<i>SACS</i>) negative	CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (<i>MPL</i>) negative
B	BARDET-BIEDL SYNDROME, ARL6-RELATED (<i>ARL6</i>) negative	CONGENITAL CHRONIC DIARRHEA (<i>DGAT1</i>) negative
	BARDET-BIEDL SYNDROME, BBS10-RELATED (<i>BBS10</i>) negative	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, ALG1-RELATED (<i>ALG1</i>) negative
	BARDET-BIEDL SYNDROME, BBS12-RELATED (<i>BBS12</i>) negative	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-Related (<i>PMM2</i>) negative
	BARDET-BIEDL SYNDROME, BBS1-RELATED (<i>BBS1</i>) negative	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (<i>MP1</i>) negative
	BARDET-BIEDL SYNDROME, BBS2-RELATED (<i>BBS2</i>) negative	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (<i>ALG6</i>) negative
	BARDET-BIEDL SYNDROME, BBS4-RELATED (<i>BBS4</i>) negative	CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (<i>SEC23B</i>) negative
	BARDET-BIEDL SYNDROME, BBS5-RELATED (<i>BBS5</i>) negative	CONGENITAL FINNISH NEPHROSIS (<i>NPHS1</i>) negative
	BARDET-BIEDL SYNDROME, BBS7-RELATED (<i>BBS7</i>) negative	CONGENITAL HYDROCEPHALUS 1 (<i>CCDC88C</i>) negative
	BARDET-BIEDL SYNDROME, BBS9-RELATED (<i>BBS9</i>) negative	CONGENITAL HYPERINSULINISM, KCNJ11-Related (<i>KCNJ11</i>) negative
	BARDET-BIEDL SYNDROME, TTC8-RELATED (<i>TTC8</i>) negative	CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (CIPA) (<i>NTRK1</i>) negative
	BARE LYMPHOCYTE SYNDROME, CIITA-RELATED (<i>CIITA</i>) negative	CONGENITAL MYASTHENIC SYNDROME, CHAT-RELATED (<i>CHAT</i>) negative
	BARTTER SYNDROME, BSND-RELATED (<i>BSND</i>) negative	CONGENITAL MYASTHENIC SYNDROME, CHRNE-RELATED (<i>CHRNE</i>) negative
	BARTTER SYNDROME, KCNJ1-RELATED (<i>KCNJ1</i>) negative	CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED (<i>COLQ</i>) negative
	BARTTER SYNDROME, SLC12A1-RELATED (<i>SLC12A1</i>) negative	CONGENITAL MYASTHENIC SYNDROME, DOK7-RELATED (<i>DOK7</i>) negative
	BATTEN DISEASE, CLN3-RELATED (<i>CLN3</i>) negative	CONGENITAL MYASTHENIC SYNDROME, RAPSN-RELATED (<i>RAPSN</i>) negative
	BETA-HEMOGLOBINOPATHIES (<i>HBB</i>) negative	CONGENITAL NEPHROTIC SYNDROME, PLCE1-RELATED (<i>PLCE1</i>) negative
	BETA-KETOTHIOLASE DEFICIENCY (<i>ACAT1</i>) negative	CONGENITAL NEUTROPENIA, G6PC3-RELATED (<i>G6PC3</i>) negative
	BETA-MANNOSIDOSIS (<i>MANBA</i>) negative	CONGENITAL NEUTROPENIA, HAX1-RELATED (<i>HAX1</i>) negative
	BETA-UREIDOPROPIONASE DEFICIENCY (<i>UPB1</i>) negative	CONGENITAL NEUTROPENIA, VPS45-RELATED (<i>VPS45</i>) negative
	BILATERAL FRONTOPIRIETAL POLYMICROGYRIA (<i>GPR56</i>) negative	CONGENITAL SECRETORY CHLORIDE DIARRHEA 1 (<i>SLC26A3</i>) negative
		CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS (<i>SLC4A11</i>) negative
		CORTICOSTERONE METHYLOXIDASE DEFICIENCY (<i>CYP11B2</i>) negative
		COSTEFF SYNDROME2 (3-METHYLGLUTACONIC ACIDURIA, TYPE 3) (<i>OPA3</i>) negative
		CRB1-RELATED RETINAL DYSTROPHIES (<i>CRB1</i>) negative
		CYSTIC FIBROSIS (<i>CFTR</i>) negative
		CYSTINOSIS (<i>CTNS</i>) negative
		CYTOCHROME C OXIDASE DEFICIENCY, PET100-RELATED (<i>PET100</i>) negative
		CYTOCHROME P450 OXIDOREDUCTASE DEFICIENCY (<i>POR</i>) negative
D	D-BIFUNCTIONAL PROTEIN DEFICIENCY (<i>HSD17B4</i>) 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 S-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) **see first page**
 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 (CTSA) **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) **negative**
 GLYCOGEN STORAGE DISEASE, TYPE 3 (AGL) **negative**
 GLYCOGEN STORAGE DISEASE, TYPE 4 (GBE1) **negative**
 GLYCOGEN STORAGE DISEASE, TYPE 7 (PFKM) **negative**

GRACILE SYNDROME (BCS1L) negativeGUANIDINOACETATE 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 (TECP2) **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 (HLCS) **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 (HLYS1) **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**

KKRABBE DISEASE (GALC) **negative****L**LAMELLAR ICHTHYOSIS, TYPE 1 (TGM1) **negative**

Patient Information

Patient Name: Donor 7820

Test Information

Ordering Physician: [REDACTED]

Clinic Information: Fairfax Cryobank



Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: 07/29/2025

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**
LIPAMIDE 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**
MEGALENCHEPHALIC 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 CbId (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 OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II (PCNT) **negative**
MICROPHthalmia / ANOPHTHALMIA, VSX2-RELATED (VSX2) **negative**
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, ACAD9-RELATED (ACAD9) **negative**
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFAF5-RELATED (NDUFAF5) **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**

M
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, TMPS53-RELATED (TMPS53) **negative**
NONSYNDROMIC INTELLECTUAL DISABILITY (CC2D1A) **negative**
NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (SAMD9) **negative**

O
OCULOCUTANEOUS ALBINISM TYPE III (TYRP1) **negative**
OCULOCUTANEOUS ALBINISM TYPE IV (SLC45A2) **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 (FBXO7) **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 7820

Test Information

Ordering Physician: [REDACTED]



Clinic Information: Fairfax Cryobank

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date:

07/29/2025

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 (SEPSEC5) **negative**
 PONTocerebellar hypoplasia, VPS53-RELATED (VPS53) **negative**
 PRIMARY CILIARY DYSPHAGIA, CCDC103-RELATED (CCDC103) **negative**
 PRIMARY CILIARY DYSPHAGIA, CCDC39-RELATED (CCDC39) **negative**
 PRIMARY CILIARY DYSPHAGIA, DNAH11-RELATED (DNAH11) **negative**
 PRIMARY CILIARY DYSPHAGIA, DNAH5-RELATED (DNAH5) **negative**
 PRIMARY CILIARY DYSPHAGIA, DNAI1-RELATED (DNAI1) **negative**
 PRIMARY CILIARY DYSPHAGIA, 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**
 PYCNODYSOSTOSIS (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 (SMARCA1) **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), FOXP1-RELATED (FOXP1) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), IKBK-RELATED (IKBK) **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, SBD5-RELATED (SBD5) **negative**
 SIALIDOSIS (NEU1) **negative**
 SJÖGREN-LARSSON SYNDROME (ALDH3A2) **negative**
 SMITH-LEMLI-OPITZ SYNDROME (DHCR7) **negative**
 SPASTIC PARAPLEGIA, TYPE 15 (ZFYZE26) **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 (WWOX) **negative**
 SPONDYLOCTOSTAL 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**
 TRICHOHYDRODYSPLASIA 1/XERODERMA PIGMENTOSUM, GROUP D (ERCC2) **negative**
 TRIMETHYLAMINURIA (FMO3) **negative**
 TRIPLE A SYNDROME (AAAS) **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 7820

Date Of Birth:

Case File ID:

Test Information

Ordering Physician:

Clinic Information: Fairfax Cryobank

Report Date: 07/29/2025



Z

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

Patient Information
Patient Name: Donor 7820

Test Information
Ordering Physician: [REDACTED]

Clinic Information: Fairfax Cryobank

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

Report Date: 07/29/2025



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, sequencing variants in exons 1-7 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. Multi-exon duplication analysis is included.

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, sequencing variants in exons 7-11 and CNV for the entire gene are not analyzed due to high sequence homology.

For GJB2, CNV analysis of upstream deletions of GJB6-D13S1830 (309kb deletion) and GJB6-D13S1854 (232kb deletion) is included.

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, sequencing variants in exons 25-29 and CNV in exons 21-29 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 7820

Test Information

Ordering Physician: [REDACTED]



Date Of Birth: [REDACTED]

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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.