



*The Trusted Choice for Donor Sperm*

## Donor 7295

### Genetic Testing Summary

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

Last Updated: 04/03/25

Donor Reported Ancestry: German, Norwegian

Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
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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.	<b>Carrier: Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency (CYP21A2) non-classic variant</b>  Negative for other genes sequenced.	Partner testing is recommended before using this donor.

\*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 7295
Date Of Birth:	[REDACTED]
Gender:	Male
Ethnicity:	Northern European Caucasian
Patient ID:	N/A
Medical Record #:	7295-[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:	12/16/2024
Sample Collected:	12/02/2024
Sample Received:	12/04/2024
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 Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency

Positive for the pathogenic variant c.844G>T (p.V282L) [Legacy name: V281L] in the CYP21A2 gene. This variant has been reported in a homozygous state or in conjunction with another variant in individual(s) with non-classic congenital adrenal hyperplasia (PMID: 19263525, 25041270, 32616876). If this individual's partner is a carrier for CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY, their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is recommended.

#### Negative for 548 out of 549 diseases

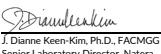
No other pathogenic variants were detected in the genes that were screened. The patient's remaining carrier risk after the negative screening results is listed for each disease/gene on the Horizon website at <https://www.natera.com/panel-option/h-all/>. Please see the following pages of this report for 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](http://naterasession.com). Clinicians with questions may contact Natera at 650-249-9090 or email [support@natera.com](mailto: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 7295

**Test Information**

Ordering Physician: [REDACTED]

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

Clinic Information: Fairfax Cryobank

Report Date: 12/16/2024

**CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY****Understanding Your Horizon Carrier Screen Results****What is Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency?**

Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency (also called 21-Hydroxylase Deficiency) is an inherited disorder that causes the adrenal glands, the organs that sit on top of the kidneys, to make decreased amounts of the hormones cortisol and aldosterone and increased amounts of male sex hormones called androgens.

There are three forms of 21-Hydroxylase Deficiency. The most common and severe form is called the 'salt-wasting type' with signs and symptoms that are often present at birth. Babies with the salt-wasting type of 21-Hydroxylase Deficiency are at risk for losing large amounts of sodium in the urine due to too low a level of aldosterone hormone. These 'salt-wasting crises' can lead to poor feeding, weight loss, dehydration, vomiting, low blood pressure, and shock, and can be life-threatening if not treated quickly. Symptoms in females include being born with external genitals that do not have the typical appearance of male or female (ambiguous genitalia). Over time, affected females may also have early puberty, rapid early growth with short adult height, increased body hair (hirsutism), male pattern baldness, irregular menstrual periods, and decreased fertility. Affected males have normal genitals at birth but are at risk for salt-wasting crises and may have increased penis size and decreased testicle size over time as well as an early growth spurt with short adult height. Some males with this form have decreased fertility due to benign growths in their testicles called 'testicular adrenal rest tumors' (TART).

The 'simple virilizing type' of 21-Hydroxylase Deficiency has similar symptoms to the salt-wasting type except babies with the simple virilizing type are not at risk for salt wasting crises.

The mildest form of 21-Hydroxylase Deficiency is called the 'non-classical type'. People with the nonclassical type of 21-Hydroxylase Deficiency have normal external genitals. Signs and symptoms may begin as early as childhood or not until adulthood and may include an early growth spurt with short adult height, early puberty, and acne. Additional symptoms in females may include excess body hair, male pattern baldness, irregular periods, and decreased fertility. Additional symptoms in males may include early and heavy facial hair and small testicles. Some people with this type never develop symptoms.

Currently, there is no cure for 21-Hydroxylase Deficiency. However, hormone replacement therapy can prevent or lessen some or all of the symptoms. Clinical trials involving potential new treatments for this condition may be available (see [www.clinicaltrials.gov](http://www.clinicaltrials.gov)).

**What causes Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency?**

21-Hydroxylase Deficiency is caused by a change, or mutation, in both copies of the CYP21A2 gene pair. These mutations cause the genes to not work properly or not work at all. The function of the CYP21A2 genes is to help make sex hormones and other hormones. When both copies of this gene do not work correctly, it leads to the symptoms described above.

21-Hydroxylase 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 CYP21A2 gene to have a child with 21-Hydroxylase Deficiency. People who are carriers for 21-Hydroxylase Deficiency are usually healthy and do not have symptoms nor do they have the disorder 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 21-Hydroxylase Deficiency, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their CYP21A2 gene mutations to the child, who will then have this condition. It is sometimes, but not always, possible to determine whether a specific mutation in the CYP21A2 gene will cause the salt-wasting type, the simple virilizing type, or the non-classic type of 21-Hydroxylase Deficiency.

Individuals found to carry more than one mutation for 21-Hydroxylase Deficiency should discuss their risk for having an affected child, and any potential effects to their own health, with their health care provider.

There are a number of other forms of Congenital Adrenal Hyperplasia, each caused by mutations in different genes. A person who is a carrier for Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency is 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.nscc.org](http://www.nscc.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 21-Hydroxylase Deficiency ordered by a health care professional. If your partner is not found to be a carrier for 21-Hydroxylase Deficiency, your risk of having an affected child is greatly reduced. Couples at risk of having a baby with 21-Hydroxylase 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 21-Hydroxylase Deficiency ordered by a health care professional. If your partner is found to be a carrier for 21-Hydroxylase Deficiency, you have several reproductive options to consider:

- Natural pregnancy with or without prenatal diagnostic testing of the fetus or testing the baby after birth for 21-Hydroxylase Deficiency
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for 21-Hydroxylase Deficiency
- Adoption or use of a sperm or egg donor who is not a carrier for 21-Hydroxylase Deficiency

**What resources are available?**

- Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/21-hydroxylase-deficiency>
- GeneReviews: <https://www.ncbi.nlm.nih.gov/books/NBK1171/>
- Prenatal diagnosis by CVS: <http://www.marchofdimes.org/chorionic-villus-sampling.aspx>

**Patient Information**

Patient Name: [REDACTED]

**Test Information**

Ordering Physician: [REDACTED]

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Clinic Information:



Report Date:

- Prenatal diagnosis by amniocentesis: <http://www.marchofdimes.org/amniocentesis.aspx>
- PGD with IVF: <http://www.natera.com/spectrum>

**Patient Information**

Patient Name: [REDACTED]

**Test Information**

Ordering Physician: [REDACTED]

Date Of Birth: [REDACTED]

Clinic Information: [REDACTED]

Case File ID: [REDACTED]

Report Date: [REDACTED]

**VARIANT DETAILS****CYP21A2, c.844G>T (p.V282L) [Legacy name: V281L], pathogenic**

- The c.844G>T (p.V282L) [Legacy name: V281L] variant in the CYP21A2 gene has been observed at a frequency of 0.5515% 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 non-classic congenital adrenal hyperplasia (PMID: 19263525, 25041270, 32616876).
- Functional studies demonstrated that this variant causes reduced enzyme activity (PMID: 24953648).
- This variant has been reported in ClinVar [ID: 12151].

**Patient Information**

Patient Name: [REDACTED]

**Test Information**

Ordering Physician: [REDACTED]



Clinic Information: [REDACTED]

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: [REDACTED]

**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 (HMGCL) negative  
3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (HADH) negative  
3-METHYLACROTONYL-CoA CARBOXYLASE 2 DEFICIENCY (MCCC2) negative  
3-PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY (PHGDH) negative

5

5-ALPHA-REDUCTASE DEFICIENCY (SRD5A2) negative

6

6-PYRUVOYL-TETRAHYDROPTERIN SYNTHASE (PTPS) DEFICIENCY (PTS) negative

A

ABCA4-RELATED CONDITIONS (ABCA4) negative  
ABETALIPOPROTEINEMIA (MTTP) negative  
ACHONDROGENESIS, TYPE 1B (SLC26A2) negative  
ACHROMATOPSY, 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 1 DEFICIENCY (ACO1) 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 (TTPA) 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, CITA-RELATED (CITA) 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-KETOThIOLASE DEFICIENCY (ACAT1) negative  
BETA-MANNOSIDOSIS (MANBA) negative  
BETA-UREIDOPROPIONASE DEFICIENCY (UPB1) negative  
BILATERAL FRONTOPARIEL POLYMICROGYRIA (GPR56) negative

BIOTINIDASE DEFICIENCY (BTD) negative

BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBDG) (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, RPRGIP1L-RELATED (RPRGIP1L) 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) see first page  
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 (MPI) 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, CHRN-RELATED (CHRN) negative  
CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED (COLQ) negative  
CONGENITAL MYASTHENIC SYNDROME, DOK7-RELATED (DOK7) negative  
CONGENITAL MYASTHENIC SYNDROME, RAPSN-RELATED (RAPSN) negative  
CONGENITAL NEPHROTIC SYNDROME, PLCE1-RELATED (PLCE1) negative  
CONGENITAL NEUTROPIA, G6PC3-RELATED (G6PC3) negative  
CONGENITAL NEUTROPIA, HAX1-RELATED (HAX1) negative  
CONGENITAL NEUTROPIA, VPS45-RELATED (VPS45) negative  
CONGENITAL SECRETORY CHLORIDE DIARRHEA 1 (SLC26A3) negative  
CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS (SLC4A11) negative  
CORTICOSTERONE METHYLOXIDASE DEFICIENCY (CYP11B2) negative  
COSTEFL SYNDROME (3-METHYGLUTAConIC 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 OXIDOREDUCTASE DEFICIENCY (POR) negative

**Patient Information**

Patient Name: [REDACTED]

**Test Information**

Ordering Physician: [REDACTED]



Clinic Information: [REDACTED]

Date Of Birth: [REDACTED]

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Report Date: [REDACTED]

**D**

D-BIFUNCTIONAL PROTEIN DEFICIENCY (HSD17B4) **negative**  
 DEAFNESS, AUTOSOMAL RECESSIVE 77 (LOXHD1) **negative**  
 DIHYDROPTERINE 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 (ADAMTS2) **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**  
 EPIPHYSAL DYSPLASIA, MULTIPLE, 7/DESBUSQUOIS 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 (IBKAP) **negative**  
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, PRF1-RELATED (PRF1) **negative**  
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STX11-RELATED (STX11) **negative**  
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STXB2P-RELATED (STXB2P) **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 (ASAHI) **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, FUC4A1-RELATED (FUC4A1) **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 OSTEODYPLASTICA (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) **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 (HLC8) **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 CALCIOSIS, 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 (INV5) **negative**  
 INFANTILE NEUROAXONAL DYSTROPHY (PLA2G6) **negative**  
 ISOLATED ECTOPIA LENTIS (ADAMTS4) **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/LARYNGOONCHOCUTANEOUS SYNDROME, LAMA3-RELATED (LAMA3) **negative**

**K**KRABBE DISEASE (GALC) **negative**

**Patient Information**

Patient Name: [REDACTED]

**Test Information**

Ordering Physician: [REDACTED]



Clinic Information: [REDACTED]

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: [REDACTED]

**L**  
 LAMELLAR ICHTHYOSIS, TYPE 1 (TGM1) negative  
 LARON SYNDROME (GHR) negative  
 LEBER CONGENITAL AMAUROSIS 2 (RPE65) negative  
 LEBER CONGENITAL AMAUROSIS TYPE A1PL1 (A1PL1) 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 (AP1S1) negative  
 MEGAENCEPHALIC 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 OSTEODYSPLOSTIC 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 (NDUFS1) negative  
 MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 2, SCO2-RELATED (SCO2) negative  
 MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 6 (COX15) 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, MFSD8-RELATED (MFSD8) 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, PIVK-RELATED (PIVK) 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 (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  
 OSTEOPENESIS IMPERFECTA TYPE VII (CRTAP) negative  
 OSTEOPENESIS IMPERFECTA TYPE VIII (P3H1) negative  
 OSTEOPENESIS IMPERFECTA TYPE XI (FKBP10) negative  
 OSTEOPENESIS 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

**Patient Information**

Patient Name: [REDACTED]

**Test Information**

Ordering Physician: [REDACTED]



Clinic Information: [REDACTED]

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: [REDACTED]

**P**POLG-RELATED DISORDERS (POLG) **negative**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 (SEPSCS) **negative**PONTOCEREBELLAR HYPOPLASIA, VP53-RELATED (VP53) **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**PSEUDOZOANTHOMA ELASTICUM (ABCC6) **negative**PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (PCD) DEFICIENCY (PCBD1) **negative**PYCNOGYROSOSTOSIS (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 IMMUNOOSSEOUS 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 POLYDACTYLY

(DYN2CH1) **negative**SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED (SBDS) **negative**SIALIDOSIS (NEU1) **negative**SJÖGREN-LARSSON SYNDROME (ALDH3A2) **negative**SMITH-LEMLI-OPITZ SYNDROME (DHCR7) **negative****Test Information**

Ordering Physician: [REDACTED]

Clinic Information: [REDACTED]

Report Date: [REDACTED]

SPASTIC PARAPLEGIA, TYPE 15 (ZFYVE26) **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**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 MEGLABLASTIC 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, SKI2-RELATED (SKI2) **negative**TRICHOHEPATOENTERIC SYNDROME, TTC37-RELATED (TTC37) **negative**TRICHOHYDROSTROPHIC 1/XERODERMA PIGMENTOSUM, GROUP D (ERCC2) **negative**TRIMETHYLAMINURIA (FMO3) **negative**TRIPLE A SYNDROME (AAAS) **negative**TSRH-RELATED CONDITIONS (TSRH) **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**

**Patient Information**

Patient Name:

**Test Information**

Ordering Physician: [REDACTED]

Date Of Birth: [REDACTED]

Clinic Information:

Case File ID: [REDACTED]

Report Date:

Z

ZELLWEGER SPECTRUM DISORDERS, PEX2-RELATED (PEX2) negative

ZELLWEGER SPECTRUM DISORDERS, PEX6-RELATED (PEX6) negative



**Patient Information**

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**Test Information**

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Date Of Birth: [REDACTED]

Clinic Information: [REDACTED]

Case File ID: [REDACTED]

Report Date: [REDACTED]

**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: [REDACTED]

**Test Information**

Ordering Physician: [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.