

SPERM DONOR GENETIC TESTING SUMMARY

Donor # 7811

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

Last Updated: 06/25/2025

Donor Reported Ancestry: Czech, English, Irish, Scottish, French, German

Jewish Ancestry: No

Genetic Test*	Result	Comments Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Expanded Genetic Disease Carrier Screening Panel attached - 549 diseases by gene sequencing and del/dup analysis.	<p>Carrier: Smith - Lemli - Opitz Syndrome (DHC7)</p> <p>Negative for other genes tested.</p>	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 7811
Date Of Birth:	[REDACTED]
Gender:	Male
Ethnicity:	Northern European Caucasian
Patient ID:	N/A
Medical Record #:	[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:	01/30/2025
Sample Collected:	01/10/2025
Sample Received:	01/13/2025
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 Smith-Lemli-Opitz Syndrome

Positive for the pathogenic variant c.976G>T (p.V326L) in the DHCR7 gene. If this individual's partner is a carrier for SMITH-LEMLI-OPITZ SYNDROME, their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

Negative for 548 out of 549 diseases

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

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

RECOMMENDATIONS

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

Christine M. Eng, M.D.
Medical Director, Baylor Genetics

Linyan Meng, Ph.D.
Laboratory Director, Baylor Genetics

J. Dianne Keen-Kim, Ph.D., FACMGG
Senior Laboratory Director, Natera

Yang Wang, Ph.D., FACMGG
Laboratory Director, Natera

Patient Information

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Date Of Birth: [REDACTED]
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**SMITH-LEMLI-OPITZ SYNDROME****Understanding Your Horizon Carrier Screen Results****What is Smith-Lemli-Opitz Syndrome?**

Smith-Lemli-Opitz (SLO) Syndrome is an inherited disorder that causes slow growth, small head size, moderate-to-severe intellectual disability, heart defects, cleft palate (opening at the roof of the mouth) and other birth defects. Lifespan in children with SLO Syndrome is often shortened and death occurs before age 2 in up to a third of affected children. The condition varies from person to person and rare individuals with SLO Syndrome have fewer symptoms and mild to no intellectual disability. Currently there is no cure for this condition and treatment is based on symptoms. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Smith-Lemli-Opitz Syndrome?

SLO Syndrome is caused by a gene change, or mutation, in both copies of the DHCR7 gene pair. These mutations cause the genes to not work properly or not work at all. The function of the DHCR7 genes is to help produce cholesterol. When there are mutations in both copies of this gene, body cells do not make enough cholesterol and toxic chemicals build up in the blood, nervous system, and other tissues and cause the symptoms described above. Increasing dietary cholesterol cannot cure SLO Syndrome and has not been proven to be helpful in improving symptoms. SLO Syndrome 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 DHCR7 gene to have a child with SLO Syndrome. People who are carriers for SLO Syndrome are usually healthy and do not have symptoms nor do they have SLO Syndrome 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 SLO Syndrome, there is a 1 in 4, or 25%, chance in each pregnancy to have a child with this condition. Individuals found to carry more than one mutation for SLO Syndrome should discuss their risk for having an affected child with their health care provider.

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). 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 SLO Syndrome ordered by a health care professional. If your partner is not found to be a carrier for SLO Syndrome, your risk of having a child with SLO Syndrome is greatly reduced. Couples at risk of having a baby with SLO Syndrome 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 SLO Syndrome ordered by a health care professional. If your partner is found to be a carrier for SLO Syndrome, 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 SLO Syndrome
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for SLO Syndrome
- Adoption or use of a sperm or egg donor who is not a carrier for SLO Syndrome

What resources are available?

- Smith-Lemli-Opitz/RSH Foundation: www.smithlemliopitz.org
- Genetics Home Reference: www.ghr.nlm.nih.gov/condition/smith-lemli-opitz-syndrome
- Prenatal diagnosis by CVS: <http://www.marchofdimes.org/chorionic-villus-sampling.aspx>
- Prenatal diagnosis by amniocentesis: <http://www.marchofdimes.org/amniocentesis.aspx>
- PGD with IVF: <http://www.natera.com/spectrum>

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Report Date: [REDACTED]

**VARIANT DETAILS****DHCR7, c.976G>T (p.V326L), pathogenic**

- The c.976G>T (p.V326L) variant in the DHCR7 gene has been observed at a frequency of 0.0017% 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 Smith-Lemli-Opitz syndrome (PMID: 9653161, 10814720, 11175299).
- This variant has been reported in ClinVar [ID: 6785].

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DISEASES SCREENED

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

Autosomal Recessive

1	17-BETA HYDROXYSTEROID DEHYDROGENASE 3 DEFICIENCY (HSD17B3) negative	BIOTINIDASE DEFICIENCY (BTD) negative
3	3-BETA-HYDROXYSTEROID DEHYDROGENASE TYPE II DEFICIENCY (HSD3B2) negative	BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBGD) (SLC19A3) negative
	3-HYDROXY-3-METHYLGLUTARYL-COENZYME A LYASE DEFICIENCY (HMGCL) negative	BLOOM SYNDROME (BLM) negative
	3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (HADH) negative	BRITTLE CORNEA SYNDROME 1 (ZNF469) negative
	3-METHYLACRYLIC ACIDURIA (MAAA) negative	BRITTLE CORNEA SYNDROME 2 (PRDM5) negative
	3-PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY (PHGDH) negative	
5	5-ALPHA-REDUCTASE DEFICIENCY (SRD5A2) negative	
6	6-PYRUVOYL-TETRAHYDROPTERIN SYNTHASE (PTPS) DEFICIENCY (PTS) negative	
A		C
	ABCA4-RELATED CONDITIONS (ABCA4) negative	CANAVAN DISEASE (ASPA) negative
	ABETALIPOPROTEINEMIA (MTPP) negative	CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (CPS1) negative
	ACHONDROGENESIS, TYPE 1B (SLC2A2) negative	CARNITINE DEFICIENCY (SLC22A5) negative
	ACHROMATOPSY, CNGB3-RELATED (CNGB3) negative	CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY (CPT1A) negative
	ACRODERMATITIS ENTEROPATHICA (SLC39A4) negative	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (CPT2) negative
	ACTION MYOCLONUS-RENAL FAILURE (AMRF) SYNDROME (SCARB2) negative	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (SLC25A20) negative
	ACUTE INFANTILE LIVER FAILURE, TRMU-RELATED (TRMU) negative	CARPENTER SYNDROME (RAB23) negative
	ACYL-COA OXIDASE I DEFICIENCY (ACOX1) negative	CARTILAGE-HAIR HYPOPLASIA (RMRP) negative
	AICARDI-GOUTIERES SYNDROME (SAMHD1) negative	CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (CASQ2) negative
	AICARDI-GOUTIERES SYNDROME, RNASEH2A-RELATED (RNASEH2A) negative	CD59-MEDIATED HEMOLYTIC ANEMIA (CD59) negative
	AICARDI-GOUTIERES SYNDROME, RNASEH2B-RELATED (RNASEH2B) negative	CEP152-RELATED MICROCEPHALY (CEP152) negative
	AICARDI-GOUTIERES SYNDROME, RNASEH2C-RELATED (RNASEH2C) negative	CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA (CEDNIK) SYNDROME (SNAP29) negative
	AICARDI-GOUTIERES SYNDROME, TREX1-RELATED (TREX1) negative	CEREBROTENDINOUS XANTHOMATOSIS (CYP27A1) negative
	ALPHA-MANNOSIDOSIS (MAN2B1) negative	CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE C (PLEKHG5) negative
	ALPHA-THALASSEMIA (HBA1/HBA2) negative	CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (NDRG1) negative
	ALPORT SYNDROME, COL4A3-RELATED (COL4A3) negative	CHEDIAK-HIGASHI SYNDROME (LYST) negative
	ALPORT SYNDROME, COL4A4-RELATED (COL4A4) negative	CHOREOACANTHOCYTOSIS (VPS13A) negative
	ALSTROM SYNDROME (ALMS1) negative	CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (CYBA) negative
	AMISH INFANTILE EPILEPSY SYNDROME (ST3GAL5) negative	CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (NCF2) negative
	ANDERMANN SYNDROME (SLC12A6) negative	CILIOPATHIES, RPGRIP1L-RELATED (RPGRIP1L) negative
	ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (GATM) negative	CITRIN DEFICIENCY (SLC25A13) negative
	ARGININEMIA (ARG1) negative	CITRULLINEMIA, TYPE 1 (ASS1) negative
	ARGINOSUCCINATE LYASE DEFICIENCY (ASL) negative	CLN10 DISEASE (CTSD) negative
	AROMATASE DEFICIENCY (CYP19A1) negative	COHEN SYNDROME (VPS13B) negative
	ASPARAGINE SYNTHETASE DEFICIENCY (ASNS) negative	COL11A2-RELATED CONDITIONS (COL11A2) negative
	ASPARTYLGLYCOSAMINURIA (AGA) negative	COMBINED MALONIC AND METHYLMALONIC ACIDURIA (ACSF3) negative
	ATAXIA WITH VITAMIN E DEFICIENCY (TTPA) negative	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (GFM1) negative
	ATAXIA-TELANGIECTASIA (ATM) negative	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (TSFM) negative
	ATAXIA-TELANGIECTASIA-LIKE DISORDER 1 (MRE11) negative	COMBINED PITUITARY HORMONE DEFICIENCY 1 (POU1F1) negative
	ATRANSFERRINEMIA (TF) negative	COMBINED PITUITARY HORMONE DEFICIENCY-2 (PROP1) negative
	AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (SLC35A3) negative	CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (CYP11B1) negative
	AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (AIRE) negative	CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (CYP17A1) negative
	AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (ARCI), SLC27A4-RELATED (SLC27A4) negative	CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (CYP21A2) negative
	AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (SACS) negative	CONGENITAL ADRENAL INSUFFICIENCY, CYP11A1-RELATED (CYP11A1) negative
B		CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (MPL) negative
	BARDET-BIEDL SYNDROME, ARL6-RELATED (ARL6) negative	CONGENITAL CHRONIC DIARRHEA (DGAT1) negative
	BARDET-BIEDL SYNDROME, BBS10-RELATED (BBS10) negative	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, ALG1-RELATED (ALG1) negative
	BARDET-BIEDL SYNDROME, BBS1-RELATED (BBS1) negative	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-Related (PMM2) negative
	BARDET-BIEDL SYNDROME, BBS2-RELATED (BBS2) negative	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (MPI) negative
	BARDET-BIEDL SYNDROME, BBS4-RELATED (BBS4) negative	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (ALG6) negative
	BARDET-BIEDL SYNDROME, BBS5-RELATED (BBS5) negative	CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (SEC23B) negative
	BARDET-BIEDL SYNDROME, BBS7-RELATED (BBS7) negative	CONGENITAL FINNISH NEPHROSIS (NPHS1) negative
	BARDET-BIEDL SYNDROME, BBS9-RELATED (BBS9) negative	CONGENITAL HYDROCEPHALUS 1 (CCDC88C) negative
	BARDET-BIEDL SYNDROME, TTC8-RELATED (TTC8) negative	CONGENITAL HYPERINSULINISM, KCNJ11-Related (KCNJ11) negative
	BARE LYMPHOCYTE SYNDROME, CITA-RELATED (CITA) negative	CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (CIPA) (NTRK1) negative
	BARTTER SYNDROME, BSND-RELATED (BSND) negative	CONGENITAL MYASTHENIC SYNDROME, CHAT-RELATED (CHAT) negative
	BARTTER SYNDROME, KCNJ1-RELATED (KCNJ1) negative	CONGENITAL MYASTHENIC SYNDROME, CHRN-RELATED (CHRN) negative
	BARTTER SYNDROME, SLC12A1-RELATED (SLC12A1) negative	CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED (COLQ) negative
	BATTEN DISEASE, CLN3-RELATED (CLN3) negative	CONGENITAL MYASTHENIC SYNDROME, DOK7-RELATED (DOK7) negative
	BETA-HEMOGLOBINOPATHIES (HBB) negative	CONGENITAL MYASTHENIC SYNDROME, RAPSN-RELATED (RAPSN) negative
	BETA-KETO THIOLASE DEFICIENCY (ACAT1) negative	CONGENITAL NEPHROTIC SYNDROME, PLCE1-RELATED (PLCE1) negative
	BETA-MANNOSIDOSIS (MANBA) negative	CONGENITAL NEUTROPENIA, G6PC3-RELATED (G6PC3) negative
	BETA-UREIDOPROPIONASE DEFICIENCY (UPB1) negative	CONGENITAL NEUTROPENIA, HAX1-RELATED (HAX1) negative
	BILATERAL FRONTOPARIEL POLYMICROGYRIA (GPR56) negative	CONGENITAL NEUTROPENIA, VPS45-RELATED (VPS45) negative
		CONGENITAL SECRETORY CHLORIDE DIARRHEA 1 (SLC26A3) negative
		CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS (SLC4A11) negative
		CORTICOSTERONE METHYLOXIDASE DEFICIENCY (CYP11B2) negative
		COSTEIFF 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
D		D-BIFUNCTIONAL PROTEIN DEFICIENCY (HSD17B4) negative

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D

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

E

EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY, CAD-RELATED (CAD) negative
 EHlers-DANLOS SYNDROME TYPE VI (PLOD1) negative
 EHlers-DANLOS SYNDROME, CLASSIC-LIKE, TNXB-RELATED (TNXB) negative
 EHlers-DANLOS SYNDROME, TYPE VII C (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
 EPIPHYSEAL DYSPLASIA, MULTIPLE, 7/DESBUQUOIS DYSPLASIA 1 (CANT1) negative
 ERCC6-RELATED DISORDERS (ERCC6) negative
 ERCC8-RELATED DISORDERS (ERCC8) negative
 ETHYLMALONIC ENCEPHALOPATHY (ETHE1) negative

F

FACTOR XI DEFICIENCY (F11) negative
 FAMILIAL DYSAUTONOMIA (IKBKA) 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 (ASA1) 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) 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 (HLCs) negative
 HOMOCYSTINURIA AND MEGALOBLASTIC ANEMIA TYPE CBLG (MTR) negative
 HOMOCYSTINURIA DUE TO DEFICIENCY OF MTHFR (MTHFR) negative
 HOMOCYSTINURIA, CBS-RELATED (CBS) negative
 HOMOCYSTINURIA, cbsE (MTRR) negative
 HYDROLETHALUS SYNDROME (HYLS1) negative
 HYPER-IGM IMMUNODEFICIENCY (CD40) negative
 HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA (HHH SYNDROME) (SLC2A15) 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 (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/LARYNGOONYCHOCUTANEOUS SYNDROME, LAMA3-RELATED (LAMA3) negative

K

KRABBE DISEASE (GALC) negative

L

LAMELLAR ICHTHYOSIS, TYPE 1 (TGM1) 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]

L

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

M

MALONYL-COA DECARBOXYLASE DEFICIENCY (MLYCD) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 1A (BCKDHA) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 1B (BCKDHB) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 2 (DBT) **negative**
 MCKUSICK-KAUFMAN SYNDROME (MKKS) **negative**
 MECKEL SYNDROME 7/NEPHRONOPHTHISIS 3 (NPHP3) **negative**
 MECKEL-GRUBER SYNDROME, TYPE 1 (MKS1) **negative**
 MECR-RELATED NEUROLOGIC DISORDER (MECR) **negative**
 MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (ACADM) **negative**
 MEDNIK SYNDROME (AP151) **negative**
 MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS (MLC1) **negative**
 MEROSIN-DEFICIENT MUSCULAR DYSTROPHY (LAMA2) **negative**
 METABOLIC ENCEPHALOPATHY AND ARRHYTHMIAS, TANGO2-RELATED (TANGO2) **negative**
 METACHROMATIC LEUKODYSTROPHY, ARSA-RELATED (ARSA) **negative**
 METACHROMATIC LEUKODYSTROPHY, PSAP-RELATED (PSAP) **negative**
 METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CBL (LMBRD1) **negative**
 METHYLMALONIC ACIDEMIA, MCEE-RELATED (MCEE) **negative**
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CBLC (MMACHC) **negative**
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CblD (MMADHC) **negative**
 METHYLMALONIC ACIDURIA, MMAA-RELATED (MMAA) **negative**
 METHYLMALONIC ACIDURIA, MMAB-RELATED (MMAB) **negative**
 METHYLMALONIC ACIDURIA, TYPE MUT(0) (MUT) **negative**
 MEVALONIC KINASE DEFICIENCY (MVK) **negative**
 MICROCEPHALIC 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 (NDUFS1) **negative**
 MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 2, SCO2-RELATED (SCO2) **negative**
 MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 6 (COX15) **negative**
 MITOCHONDRIAL DNA DEPLETION SYNDROME 2 (TK2) **negative**

MITOCHONDRIAL DNA DEPLETION SYNDROME 3 (DGUOK) **negative**MITOCHONDRIAL MYOPATHY AND SIDEROBLASTIC ANEMIA (MLASA1) (PUS1) **negative**
 MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (HADHB) **negative**MOLYBDENUM COFACTOR DEFICIENCY TYPE B (MOC52) **negative**MOLYBDENUM COFACTOR DEFICIENCY, TYPE A (MOC51) **negative**MUCOLIPIDOSIS II/III A (GNPTAB) **negative**MUCOLIPIDOSIS III GAMMA (GNPTG) **negative**MUCOLIPIDOSIS, TYPE IV (MCOLN1) **negative**MUCOPOLYSACCHARIDOSIS, TYPE I (HURLER SYNDROME) (IDUA) **negative**MUCOPOLYSACCHARIDOSIS, TYPE III A (SANFILIPPO A) (SGSH) **negative**MUCOPOLYSACCHARIDOSIS, TYPE III B (SANFILIPPO B) (NAGLU) **negative**MUCOPOLYSACCHARIDOSIS, TYPE III C (SANFILIPPO C) (HGSNAT) **negative**MUCOPOLYSACCHARIDOSIS, TYPE III D (SANFILIPPO D) (GNS) **negative**MUCOPOLYSACCHARIDOSIS, TYPE IV A (MORQUO SYNDROME) (GALNS) **negative**MUCOPOLYSACCHARIDOSIS, TYPE IV B/GM1 GANGLIOSIDOSIS (GLB1) **negative**MUCOPOLYSACCHARIDOSIS, TYPE IX (HYAL1) **negative**MUCOPOLYSACCHARIDOSIS, TYPE VI (MAROTEAUX-LAMY) (ARSB) **negative**MUCOPOLYSACCHARIDOSIS, TYPE VII (GUSB) **negative**MULIBREY NANISM (TRIM37) **negative**MULTIPLE PTERYGIUM SYNDROME, CHRNG-RELATED/ESCOBAR SYNDROME (CHRNG) **negative**MULTIPLE SULFATASE DEFICIENCY (SUMF1) **negative**MUSCLE-EYE-BRAIN DISEASE, POMGNT1-RELATED (POMGNT1) **negative**MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (RXYLT1) **negative**MUSK-RELATED CONGENITAL MYASTHENIC SYNDROME (MUSK) **negative**MYONEUROGASTROINTESTINAL ENCEPHALOPATHY (MNGIE) (TYMP) **negative**MYOTONIA CONGENITA (CLCN1) **negative****N**N-ACETYLGUTAMATE 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, PJVK-RELATED (PJVK) **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**NONMOPHOSPHATEMIC TUMORAL CALCINOSIS (SAMD9) **negative****O**OCULOLOCUTANEOUS ALBINISM TYPE III (TYRP1) **negative**OCULOLOCUTANEOUS ALBINISM TYPE IV (SLC45A2) **negative**OCULOLOCUTANEOUS ALBINISM, OCA2-RELATED (OCA2) **negative**OCULOLOCUTANEOUS 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 LEFEVRE 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: [REDACTED]

Test Information

Ordering Physician: [REDACTED]



Clinic Information: [REDACTED]

Date Of Birth: [REDACTED]

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Report Date: [REDACTED]

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 (SEPSECS) negative
 PONTOCEREBELLAR HYPOPLASIA, VPS53-RELATED (VPS53) negative
 PRIMARY CILIARY DYSKINESIA, CCDC103-RELATED (CCDC103) negative
 PRIMARY CILIARY DYSKINESIA, CCDC39-RELATED (CCDC39) negative
 PRIMARY CILIARY DYSKINESIA, DNAH11-RELATED (DNAH11) negative
 PRIMARY CILIARY DYSKINESIA, DNAH5-RELATED (DNAH5) negative
 PRIMARY CILIARY DYSKINESIA, DNA11-RELATED (DNA11) negative
 PRIMARY CILIARY DYSKINESIA, DNA12-RELATED (DNA12) 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
 PYCNOYDYSOSTOSIS (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
 RLRP1-RELATED RETINOPATHY (RLRP1) negative
 ROBERTS SYNDROME (ESCO2) negative
 RYR1-RELATED CONDITIONS (RYR1) negative

S

SALLA DISEASE (SLC17A5) negative
 SANDHOFF DISEASE (HEXB) negative
 SCHIMKE IMMUNOSSEOUS DYSPLASIA (SMARCAL1) negative
 SCHINDLER DISEASE (NAGA) negative
 SEGAWA SYNDROME, TH-RELATED (TH) negative
 SENIOR-LOKEN SYNDROME 4/NEPHRONOPHTHISIS 4 (NPHP4) negative
 SEPIAFTERIN 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
 (DYNCH2H1) negative
 SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED (SBDS) negative
 SIALIDOSIS (NEU1) negative
 SJÖGREN-LARSSON SYNDROME (ALDH3A2) negative
 SMITH-LEMLI-OPITZ SYNDROME (DHCR7) see first page
 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
 SPONDYLOCOLSTAL 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
 TRICHOHYDROSTROPHY 1/XERODERMA PIGMENTOSUM, GROUP D (ERCC2) negative
 TRIMETHYLMALINURIA (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:

Test Information

Ordering Physician: [REDACTED]

Date Of Birth: [REDACTED]

Clinic Information:

Case File ID: [REDACTED]

Report Date:

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ZELLWEGER SPECTRUM DISORDERS, PEX6-RELATED (PEX6) negative



Patient Information

Patient Name: [REDACTED]

Test Information

Ordering Physician: [REDACTED]

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

Clinic Information: [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

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Test Information

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Report Date: [REDACTED]

**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.