
SPERM DONOR GENETIC TESTING SUMMARY**Donor # 7185**

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

Last Updated: 8/22/2025

Donor Reported Ancestry: Cuban, Spanish

Jewish Ancestry: No

Genetic Test*	Result	Comments Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobinopathy 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.	Negative for other genes tested.	

*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 7185
Date Of Birth:	[REDACTED]
Gender:	Male
Ethnicity:	Hispanic/Latin American
Patient ID:	N/A
Medical Record #:	N/A
Collection Kit:	[REDACTED]
Accession ID:	N/A
Case File ID:	[REDACTED]

Test Information	
Ordering Physician:	[REDACTED]
Clinic Information:	Fairfax Cryobank
Phone:	[REDACTED]
Report Date:	06/25/2025
Sample Collected:	06/11/2025
Sample Received:	06/12/2025
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:



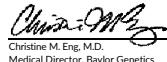
Negative for 549 out of 549 diseases

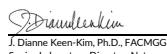
No Pathogenic variants were detected in the genes that were screened. The patient's remaining carrier risk after the negative screening results is listed for each disease/gene on the Horizon website at <https://www.natera.com/panel-option/h-all/>. Please see the following pages of this report for a comprehensive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


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Patient Information

Patient Name: Donor 7185

Test Information

Ordering Physician: [REDACTED]



Clinic Information: Fairfax Cryobank

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: 06/25/2025

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-METHYLACRYLONYL-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 (MTPP) negative
ACHONDROGENESIS, TYPE 1B (SLC2A2) negative
ACHROMATOPSIA, CNGB3-RELATED (CNGB3) negative
ACRODERMATITIS ENTEROPATHICA (SLC39A4) negative
ACTION MYOCLONUS-RENAL FAILURE (AMRF) SYNDROME (SCARB2) negative
ACUTE INFANTILE LIVER FAILURE, TRMU-RELATED (TRMU) negative
ACYL-COA OXIDASE I DEFICIENCY (ACOX1) negative
AICARDI-GOUTIERES SYNDROME (SAMHD1) negative
AICARDI-GOUTIERES SYNDROME, RNASEH2A-RELATED (RNASEH2A) negative
AICARDI-GOUTIERES SYNDROME, RNASEH2B-RELATED (RNASEH2B) negative
AICARDI-GOUTIERES SYNDROME, RNASEH2C-RELATED (RNASEH2C) negative
AICARDI-GOUTIERES SYNDROME, TREX1-RELATED (TREX1) negative
ALPHA-MANNOSIDOSIS (MAN2B1) negative
ALPHA-THALASSEMIA (HBA1/HBA2) negative
ALPORT SYNDROME, COL4A3-RELATED (COL4A3) negative
ALPORT SYNDROME, COL4A4-RELATED (COL4A4) negative
ALSTROM SYNDROME (ALMS1) negative
AMISH INFANTILE EPILEPSY SYNDROME (ST3GAL5) negative
ANDERMANN SYNDROME (SLC12A6) negative
ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (GATM) negative
ARGININEMIA (ARG1) negative
ARGINOSUCCINATE 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-KETO THIOLASE 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 (BTBD) (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-ACYL CARNITINE TRANSLOCASE DEFICIENCY (SLC25A20) negative
CARPENTER SYNDROME (RAB23) negative
CARTILAGE-HAIR HYPOPLASIA (RMRP) negative
CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (CASQ2) negative
CD59-MEDIATED HEMOLYTIC ANEMIA (CD59) negative
CEP152-RELATED MICROCEPHALY (CEP152) negative
CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA (CEDNIK) SYNDROME (SNAP29) negative
CEREBROTENDINOUS XANTHOMATOSIS (CYP27A1) negative
CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE C (PLEKHG5) negative
CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (NDRG1) negative
CHEDIAK-HIGASHI SYNDROME (LYST) negative
CHOREOACANTHOCYTOSIS (VPS13A) negative
CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (CYBA) negative
CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (NCF2) negative
CILIOPATHIES, RPGRIP1L-RELATED (RPGRIP1L) negative
CITRIN DEFICIENCY (SLC25A13) negative
CITRULLINEMIA, TYPE 1 (ASS1) negative
CLN10 DISEASE (CTSD) negative
COHEN SYNDROME (VPS13B) negative
COL11A2-RELATED CONDITIONS (COL11A2) negative
COMBINED MALONIC AND METHYLMALONIC ACIDURIA (ACSF3) negative
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (GFM1) negative
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (TSFM) negative
COMBINED PITUITARY HORMONE DEFICIENCY 1 (POU1F1) negative
COMBINED PITUITARY HORMONE DEFICIENCY-2 (PROP1) negative
CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (CYP11B1) negative
CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (CYP17A1) negative
CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (CYP21A2) negative
CONGENITAL ADRENAL INSUFFICIENCY, CYP11A1-RELATED (CYP11A1) negative
CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (MPL) negative
CONGENITAL CHRONIC DIARRHEA (DGAT1) negative
CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, ALG1-RELATED (ALG1) negative
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-Related (PMM2) negative
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (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 NEUTROPENIA, G6PC3-RELATED (G6PC3) negative
CONGENITAL NEUTROPENIA, HAX1-RELATED (HAX1) negative
CONGENITAL NEUTROPENIA, VPS45-RELATED (VPS45) negative
CONGENITAL SECRETORY CHLORIDE DIARRHEA 1 (SLC26A3) negative
CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS (SLC4A11) negative
CORTICOSTERONE METHYLOXIDASE DEFICIENCY (CYP11B2) negative
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

D

D-BIFUNCTIONAL PROTEIN DEFICIENCY (HSD17B4) negative

Patient Information

Patient Name: [REDACTED]

Test Information

Ordering Physician: [REDACTED]



Clinic Information: [REDACTED]

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Report Date: [REDACTED]

D

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
 EHRLERS-DANLOS SYNDROME TYPE VI (PLOD1) negative
 EHRLERS-DANLOS SYNDROME, CLASSIC-LIKE, TNXB-RELATED (TNXB) negative
 EHRLERS-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 (IBKAP) negative
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, PRF1-RELATED (PRF1) negative
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STX11-RELATED (STX11) negative
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STXBP2-RELATED (STXBP2) negative
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, UNC13D-RELATED (UNC13D) negative
 FAMILIAL HYPERCHOLESTEROLEMIA, LDLRAP1-RELATED (LDLRAP1) negative
 FAMILIAL HYPERCHOLESTEROLEMIA, LDLR-RELATED (LDLR) negative
 FAMILIAL HYPERINSULINISM, ABCC8-RELATED (ABCC8) negative
 FAMILIAL NEPHROGENIC DIABETES INSIPIDUS, AQP2-RELATED (AQP2) negative
 FANCONI ANEMIA, GROUP A (FANCA) negative
 FANCONI ANEMIA, GROUP C (FANCC) negative
 FANCONI ANEMIA, GROUP D2 (FANCD2) negative
 FANCONI ANEMIA, GROUP E (FANCE) negative
 FANCONI ANEMIA, GROUP F (FANCF) negative
 FANCONI ANEMIA, GROUP G (FANCG) negative
 FANCONI ANEMIA, GROUP I (FANCI) negative
 FANCONI ANEMIA, GROUP J (BRIP1) negative
 FANCONI ANEMIA, GROUP L (FANCL) negative
 FARBER LIPOGRANULOMATOSIS (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
 FUROSISIDOSIS, 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 (GTS) 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 PARAPARESIS, CYP7B1-RELATED (CYP7B1) negative
 HERMANSKY-PUDLAK SYNDROME, AP3B1-RELATED (AP3B1) negative
 HERMANSKY-PUDLAK SYNDROME, BLOC1S3-RELATED (BLOC1S3) negative
 HERMANSKY-PUDLAK SYNDROME, BLOC1S6-RELATED (BLOC1S6) negative
 HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED (HPS1) negative
 HERMANSKY-PUDLAK SYNDROME, HPS3-RELATED (HPS3) negative
 HERMANSKY-PUDLAK SYNDROME, HPS4-RELATED (HPS4) negative
 HERMANSKY-PUDLAK SYNDROME, HPS5-RELATED (HPS5) negative
 HERMANSKY-PUDLAK SYNDROME, HPS6-RELATED (HPS6) negative
 HOLOCARBOXYLASE SYNTHETASE DEFICIENCY (HLCs) negative
 HOMOCYSTINURIA AND MEGALOBLASTIC ANEMIA TYPE CBLG (MTR) negative
 HOMOCYSTINURIA DUE TO DEFICIENCY OF MTHFR (MTHFR) negative
 HOMOCYSTINURIA, CBS-RELATED (CBS) negative
 HOMOCYSTINURIA, Type cb1e (MTRR) negative
 HYDROLETHALUS SYNDROME (HYLS1) negative
 HYPER-IGM IMMUNODEFICIENCY (CD40) negative
 HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA (HHH SYNDROME) (SLC25A15) negative
 HYPERPHOSPHATEMIC FAMILIAL TUMORAL CALCINOSIS, GALNT3-RELATED (GALNT3) negative
 HYPOMYELINATING LEUKODYSTROPHY 12 (VPS11) negative
 HYPOPHOSPHATASIA, ALPL-RELATED (ALPL) negative

I

IMERSLUND-GRÄSBECK SYNDROME 2 (AMN) negative
 IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES (ICF) SYNDROME, DNMT3B-RELATED (DNMT3B) negative
 IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES (ICF) SYNDROME, ZBTB24-RELATED (ZBTB24) negative
 INCLUSION BODY MYOPATHY 2 (GNE) negative
 INFANTILE CEREBRAL AND CEREBELLAR ATROPHY (MED17) negative
 INFANTILE NEPHRONOPHTHISIS (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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L
 LARON SYNDROME (GHR) **negative**
 LEBER CONGENITAL AMAUROSIS 2 (RPE65) **negative**
 LEBER CONGENITAL AMAUROSIS TYPE AIPL1 (AIPL1) **negative**
 LEBER CONGENITAL AMAUROSIS TYPE GUCY2D (GUCY2D) **negative**
 LEBER CONGENITAL AMAUROSIS TYPE TULP1 (TULP1) **negative**
 LEBER CONGENITAL AMAUROSIS, IQCB1-RELATED/SENIOR-LOKEN SYNDROME 5 (IQCB1) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE CEP290 (CEP290) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE LCA5 (LCA5) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE RDH12 (RDH12) **negative**
 LEIGH SYNDROME, FRENCH-CANADIAN TYPE (LRPPRC) **negative**
 LETHAL CONGENITAL CONTRACTURE SYNDROME 1 (GLE1) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER (EIF2B5) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B1-RELATED (EIF2B1) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B2-RELATED (EIF2B2) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B3-RELATED (EIF2B3) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B4-RELATED (EIF2B4) **negative**
 LIG4 SYNDROME (LIG4) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 8 (TRIM32) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2A (CAPN3) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2B (DYSF) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2C (SGCG) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2D (SGCA) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2E (SGCB) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2F (SGCD) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2I (FKRP) **negative**
 LIPOAMIDE DEHYDROGENASE DEFICIENCY (DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY) (DLD) **negative**
 LIPOID ADRENAL HYPERPLASIA (STAR) **negative**
 LIPOPROTEIN LIPASE DEFICIENCY (LPL) **negative**
 LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (HADHA) **negative**
 LRAT-RELATED CONDITIONS (LRAT) **negative**
 LUNG DISEASE, IMMUNODEFICIENCY, AND CHROMOSOME BREAKAGE SYNDROME (LICS) (NSMCE3) **negative**
 LYSINURIC PROTEIN INTOLERANCE (SLC7A7) **negative**

M
 MALONYL-COA DECARBOXYLASE DEFICIENCY (MLYCD) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 1A (BCKDHA) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 1B (BCKDHB) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 2 (DBT) **negative**
 MCKUSICK-KAUFMAN SYNDROME (MKKS) **negative**
 MECKEL SYNDROME 7/NEPHRONOPHTHISIS 3 (NPHP3) **negative**
 MECKEL-GRUBER SYNDROME, TYPE 1 (MKS1) **negative**
 MECR-RELATED NEUROLOGIC DISORDER (MECR) **negative**
 MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (ACADM) **negative**
 MEDNIK SYNDROME (AP151) **negative**
 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 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**

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**
 NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (SAMD9) **negative**

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

P
 PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION (PANK2) **negative**
 PAPILLON 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

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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
 PYCNOHYDROSIS (*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 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
 (DYNCH2H1) negative
 SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED (*SBDS*) negative
 SIALIDOSIS (*NEU1*) negative
 SJÖGREN-LARSSON SYNDROME (*ALDH3A2*) negative
 SMITH-LEMLI-OPITZ SYNDROME (*DHCR7*) negative
 SPASTIC PARAPLEGIA, TYPE 15 (*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 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 (*TNC2*) negative

TRICHOHEPATOENTERIC SYNDROME, SKIC2-RELATED (*SKIC2*) negative

TRICHOHEPATOENTERIC SYNDROME, TTC37-RELATED (*TTC37*) negative

TRICHOHYDROSTROPHY 1/XERODERMA PIGMENTOSUM, GROUP D (*ERCC2*) negative

TRIMETHYLAMINURIA (*FMO3*) negative

TRIPLE A SYNDROME (*AAAS*) negative

TSHR-RELATED CONDITIONS (*TSHR*) negative

TYROSINEMIA TYPE III (*HPD*) negative

TYROSINEMIA, TYPE 1 (*FAH*) negative

TYROSINEMIA, TYPE 2 (*TAT*) negative

U

USHER SYNDROME, TYPE 1B (*MYO7A*) negative

USHER SYNDROME, TYPE 1C (*USH1C*) negative

USHER SYNDROME, TYPE 1D (*CDH23*) negative

USHER SYNDROME, TYPE 1F (*PCDH15*) negative

USHER SYNDROME, TYPE 1J/DEAFNESS, AUTOSOMAL RECESSIVE, 48 (*CIB2*) negative

USHER SYNDROME, TYPE 2A (*USH2A*) negative

USHER SYNDROME, TYPE 2C (*ADGRV1*) negative

USHER SYNDROME, TYPE 3 (*CLRN1*) negative

V

VERY LONG-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (*ACADVL*) negative

VICI SYNDROME (*EPG5*) negative

VITAMIN D-DEPENDENT RICKETS, TYPE 1A (*CYP27B1*) negative

VITAMIN D-RESISTANT RICKETS TYPE 2A (*VDR*) negative

VLDLR-ASSOCIATED CEREBELLAR HYPOPLASIA (*VLDLR*) negative

W

WALKER-WARBURG SYNDROME, CRPPA-RELATED (*CRPPA*) negative

WALKER-WARBURG SYNDROME, FKTN-RELATED (*FKTN*) negative

WALKER-WARBURG SYNDROME, LARGE1-RELATED (*LARGE1*) negative

WALKER-WARBURG SYNDROME, POMT1-RELATED (*POMT1*) negative

WALKER-WARBURG SYNDROME, POMT2-RELATED (*POMT2*) negative

WARSAW BREAKAGE SYNDROME (*DDX11*) negative

WERNER SYNDROME (*WRN*) negative

WILSON DISEASE (*ATP7B*) negative

WOLCOTT-RALLISON SYNDROME (*EIF2AK3*) negative

WOLMAN DISEASE (*LIPA*) negative

WOODHOUSE-SAKATI SYNDROME (*DCAF17*) negative

X

XERODERMA PIGMENTOSUM VARIANT TYPE (*POLH*) negative

XERODERMA PIGMENTOSUM, GROUP A (*XPA*) negative

XERODERMA PIGMENTOSUM, GROUP C (*XPC*) negative

Z

ZELLWEGER SPECTRUM DISORDER, PEX13-RELATED (*PEX13*) negative

ZELLWEGER SPECTRUM DISORDER, PEX16-RELATED (*PEX16*) negative

ZELLWEGER SPECTRUM DISORDER, PEX5-RELATED (*PEX5*) negative

ZELLWEGER SPECTRUM DISORDERS, PEX10-RELATED (*PEX10*) negative

ZELLWEGER SPECTRUM DISORDERS, PEX12-RELATED (*PEX12*) negative

ZELLWEGER SPECTRUM DISORDERS, PEX1-RELATED (*PEX1*) negative

ZELLWEGER SPECTRUM DISORDERS, PEX26-RELATED (*PEX26*) negative

ZELLWEGER SPECTRUM DISORDERS, PEX2-RELATED (*PEX2*) negative

Patient Information

Patient Name:

Test Information

Ordering Physician: [REDACTED]

Date Of Birth: [REDACTED]

Clinic Information:

Case File ID: [REDACTED]

Report Date:

Z

ZELLWEGER SPECTRUM DISORDERS, PEX6-RELATED (PEX6) negative



Patient Information

Patient Name: [REDACTED]

Test Information

Ordering Physician: [REDACTED]

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]



Clinic Information: [REDACTED]

Date Of Birth: [REDACTED]

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Spinal Muscular Atrophy (SMN1)

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

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

Variant Classification

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

Negative Results

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

Additional Comments

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