

GENETIC TESTING SUMMARY

Sperm Donor # 8416

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

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

Last Updated: 09/08/2026

Genetic Test	Result	Comments
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Expanded Genetic Carrier Screening: 549 diseases by gene sequencing and del/dup analysis.	Carrier: POLG-Related Disorders (POLG) Negative for other genes tested.	Egg source carrier screening is indicated before using this donor

Patient Information

Patient Name: Donor 8416

Date Of Birth: [REDACTED]

Gender: Male

Patient ID: N/A

Medical Record #: N/A

Collection Kit: [REDACTED]

Accession ID: N/A

Case File ID: [REDACTED]

Ethnicity: Other

Test Information

Ordering Physician: [REDACTED]

Clinic Information: Fairfax Cryobank

Phone: [REDACTED]

Report Date: 03/04/2026

Sample Collected: 02/23/2026

Sample Received: 02/24/2026

Sample Type: Blood

**Horizon™**
Advanced carrier screening**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 POLG-Related Disorders**

Positive for the pathogenic variant c.2246T>C (p.F749S) in the POLG gene. Although most variants in this gene are associated with an autosomal recessive form of POLG-Related Disorders, some rare POLG variants may cause an autosomal dominant form of the condition. To our knowledge, there is insufficient evidence that this variant causes an autosomal dominant form of this condition. If this individual's partner is a carrier for POLG-RELATED DISORDERS, their chance to have a child with this condition is likely 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

Negative for 548 out of 549 diseases

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

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

RECOMMENDATIONS

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

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Patient Information

Patient Name: Donor 8416

Test Information

Ordering Physician: [REDACTED]



Horizon™
Advanced carrier screening

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Clinic Information: Fairfax Cryobank

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POLG-RELATED DISORDERS

Understanding Your Horizon Carrier Screen Results

What is POLG-Related Disorders?

POLG-Related Disorders are a group of related inherited disorders that affect the muscles, nerves, and brain. The POLG-Related Disorders include: Alpers-Huttenlocher Syndrome (AHS); Ataxia Neuropathy Spectrum (ANS); Childhood Myocerebrohepatopathy Spectrum (MCHS); Myoclonic Epilepsy Myopathy Sensory Ataxia (MEMSA); and Progressive External Ophthalmoplegia. Age at the start of symptoms varies from infancy to adulthood.

- Alpers-Huttenlocher Syndrome (AHS) is the most serious of the POLG-Related Disorders and causes severe inflammation and damage to the brain (encephalopathy) along with chronic, poorly-controlled seizures, and progressive liver damage. Other symptoms may include loss of developmental skills, coordination and balance problems, abnormal movements, absent reflexes, poor muscle tone (hypotonia), neuropathy (pain in the arms and legs), and/or loss of vision and hearing.
- Ataxia Neuropathy Spectrum (ANS) causes movement and coordination problems along with seizures and neuropathy (numbness and pain in the arms and legs). Some individuals with ANS have muscle twitches and/or vision loss. About half of individuals with ANS have weakness of the eye muscles (ophthalmoplegia) and drooping eyelids (ptosis).
- Childhood Myocerebrohepatopathy Spectrum (MCHS) symptoms start between infancy and 3 years of age and include developmental delay, muscle weakness, and poor growth. Some affected children have severe liver and kidney disease, pancreatitis, episodes of vomiting and/or hearing loss.
- Myoclonic Epilepsy Myopathy Sensory Ataxia (MEMSA) causes seizures, muscle weakness, and movement and coordination problems (ataxia), usually starting in early adulthood.
- Progressive External Ophthalmoplegia, Autosomal Recessive has symptoms that include weakness of the eye muscles that worsens with time and causes drooping of the eyelids (ptosis) and paralysis of the outer muscles that move the eye. Symptoms typically start in early-to-mid adulthood and are usually limited to the symptoms above, although some affected individuals develop some of the symptoms seen in the above disorders over time.
- Progressive External Ophthalmoplegia, Autosomal Dominant has symptoms similar to the autosomal recessive form but also include muscle weakness and fatigue, especially during exercise. Additional symptoms of the dominant form in some affected individuals include hearing loss, neuropathy (numbness and pain in the limbs), coordination and balance problems, and cataracts. Affected individuals may have an increased risk for Parkinson Disease.
- There is no cure for any of these disorders and treatment is based on symptoms. Clinical trials involving potential new treatments for these conditions may be available (see www.clinicaltrials.gov).

What causes POLG-Related Disorders?

POLG-Related Disorders are caused by a change, or mutation, in both copies of the POLG gene pair. These mutations cause the genes to not work properly or not work at all. When both copies of this gene do not work correctly, it leads to the symptoms of one of the disorders described above. It is sometimes, but not always, possible to determine which of the POLG-Related Disorders a specific mutation in the POLG gene will cause. All of the POLG-Related Disorders except one are 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 POLG gene to have a child with one of these conditions. People who are carriers for POLG-Related Disorders are usually healthy and do not typically have these disorders 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 POLG-Related Disorders, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their POLG gene mutations to the child, who will then have one of these disorders. Individuals found to carry more than one mutation for POLG-Related Disorders should discuss their risk for having an affected child, and any potential risks to their own health, with their health care provider. Less commonly, Progressive External Ophthalmoplegia is inherited in an autosomal dominant manner, usually from an affected parent. This means that a person with a mutation in one copy of the POLG gene will be affected and have symptoms of Progressive External Ophthalmoplegia, Autosomal Dominant. Adults with the autosomal dominant form of this disorder have a 1 in 2, or 50%, chance in each pregnancy of passing on the mutation to a child, who would then be affected with Progressive External Ophthalmoplegia, Autosomal Dominant. It is sometimes, but not always, possible to determine whether a specific mutation in the POLG gene will cause the autosomal recessive or autosomal dominant form of this condition (or one of the other POLG-Related disorders).

What can I do next?

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website (www.nsgc.org). Your siblings and other relatives are at increased risk to also have this mutation. You are encouraged to inform your family members of your test results as they may wish to consider being tested themselves. If you are pregnant, your partner can have carrier screening for POLG-Related Disorders ordered by a health care professional. If your partner is not found to be a carrier, your risk of having a child with one of these conditions is greatly reduced. Couples at risk of having a baby with a POLG-Related Disorder 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. If you are not yet pregnant, your partner can have carrier screening for POLG-Related Disorders ordered by a health care professional. If your partner is found to be a carrier for POLG-Related Disorders, you have several reproductive options to consider:

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

What resources are available?

- Genetics Home Reference: <https://ghr.nlm.nih.gov/gene/POLG>
- GeneReviews: <https://www.ncbi.nlm.nih.gov/books/NBK26471/>



Patient Information

Patient Name: Donor 8416

Test Information

Ordering Physician: [REDACTED]



Horizon™
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Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Clinic Information: Fairfax Cryobank

Report Date: 03/04/2026

- Prenatal diagnosis done through CVS: <http://www.marchofdimes.org/chorionic-villus-sampling.aspx>
- Prenatal diagnosis done through Amniocentesis: <http://www.marchofdimes.org/amniocentesis.aspx>
- PGD with IVF: <http://www.natera.com/spectrum>
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VARIANT DETAILS**POLG, c.2246T>C (p.F749S), pathogenic**

- The c.2246T>C (p.F749S) variant in the POLG gene has been observed at a frequency of 0.0064% 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 POLG-related disorders (PMID: 16545482, 21880868).
- This variant has been reported in ClinVar [ID: 206520].

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DISEASES SCREENED

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

Autosomal Recessive**1**17-BETA HYDROXYSTEROID DEHYDROGENASE 3 DEFICIENCY (*HSD17B3*) **negative****3**

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

55-ALPHA-REDUCTASE DEFICIENCY (*SRD5A2*) **negative****6**6-PYRUVYL-TETRAHYDROPTERIN SYNTHASE (*PTPS*) DEFICIENCY (*PTS*) **negative****A**

ABCA4-RELATED CONDITIONS (*ABCA4*) **negative**
 ABETALIPROTEINEMIA (*MTTP*) **negative**
 ACHONDROGENESIS, TYPE 1B (*SLC26A2*) **negative**
 ACHROMATOPSIA, CNGB3-RELATED (*CNGB3*) **negative**
 ACRODERMATITIS ENTEROPATHICA (*SLC39A4*) **negative**
 ACTION MYOCLONUS-RENAL FAILURE (AMRF) SYNDROME (*SCARB2*) **negative**
 ACUTE INFANTILE LIVER FAILURE, TRMU-RELATED (*TRMU*) **negative**
 ACYL-COA OXIDASE I DEFICIENCY (*ACOX1*) **negative**
 AICARDI-GOUTIERES SYNDROME (*SAMHD1*) **negative**
 AICARDI-GOUTIERES SYNDROME, RNASEH2A-RELATED (*RNASEH2A*) **negative**
 AICARDI-GOUTIERES SYNDROME, RNASEH2B-RELATED (*RNASEH2B*) **negative**
 AICARDI-GOUTIERES SYNDROME, RNASEH2C-RELATED (*RNASEH2C*) **negative**
 AICARDI-GOUTIERES SYNDROME, TREX1-RELATED (*TREX1*) **negative**
 ALPHA-MANNOSIDOSIS (*MAN2B1*) **negative**
 ALPHA-THALASSEMIA (*HBA1/HBA2*) **negative**
 ALPORT SYNDROME, COL4A3-RELATED (*COL4A3*) **negative**
 ALPORT SYNDROME, COL4A4-RELATED (*COL4A4*) **negative**
 ALSTROM SYNDROME (*ALMS1*) **negative**
 AMISH INFANTILE EPILEPSY SYNDROME (*ST3GAL5*) **negative**
 ANDERMANN SYNDROME (*SLC12A6*) **negative**
 ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (*GATM*) **negative**
 ARGININEMIA (*ARG1*) **negative**
 ARGININOSUCCINATE LYASE DEFICIENCY (*ASL*) **negative**
 AROMATASE DEFICIENCY (*CYP19A1*) **negative**
 ASPARAGINE SYNTHETASE DEFICIENCY (*ASNS*) **negative**
 ASPARTYLGLYCOSAMINURIA (*AGA*) **negative**
 ATAXIA WITH VITAMIN E DEFICIENCY (*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**

BBARDET-BIEDL SYNDROME, ARL6-RELATED (*ARL6*) **negative**

BARDET-BIEDL SYNDROME, BBS10-RELATED (*BBS10*) **negative**
 BARDET-BIEDL SYNDROME, BBS12-RELATED (*BBS12*) **negative**
 BARDET-BIEDL SYNDROME, BBS1-RELATED (*BBS1*) **negative**
 BARDET-BIEDL SYNDROME, BBS2-RELATED (*BBS2*) **negative**
 BARDET-BIEDL SYNDROME, BBS4-RELATED (*BBS4*) **negative**
 BARDET-BIEDL SYNDROME, BBS5-RELATED (*BBS5*) **negative**
 BARDET-BIEDL SYNDROME, BBS7-RELATED (*BBS7*) **negative**
 BARDET-BIEDL SYNDROME, BBS9-RELATED (*BBS9*) **negative**
 BARDET-BIEDL SYNDROME, TTC8-RELATED (*TTC8*) **negative**
 BARE LYMPHOCYTE SYNDROME, CIITA-RELATED (*CIITA*) **negative**
 BARTTER SYNDROME, BSND-RELATED (*BSND*) **negative**
 BARTTER SYNDROME, KCNJ1-RELATED (*KCNJ1*) **negative**
 BARTTER SYNDROME, SLC12A1-RELATED (*SLC12A1*) **negative**
 BATTEN DISEASE, CLN3-RELATED (*CLN3*) **negative**
 BETA-HEMOGLOBINOPATHIES (*HBB*) **negative**
 BETA-KETOTHIOLASE DEFICIENCY (*ACAT1*) **negative**
 BETA-MANNOSIDOSIS (*MANBA*) **negative**
 BETA-UREIDOPROPIONASE DEFICIENCY (*UPB1*) **negative**
 BILATERAL FRONTOPIRIETAL POLYMICROGYRIA (*GPR56*) **negative**
 BIOTINIDASE DEFICIENCY (*BTD*) **negative**
 BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBGD) (*SLC19A3*) **negative**
 BLOOM SYNDROME (*BLM*) **negative**
 BRITTLE CORNEA SYNDROME 1 (*ZNF469*) **negative**
 BRITTLE CORNEA SYNDROME 2 (*PRDM5*) **negative**

C

CANAVAN DISEASE (*ASPA*) **negative**
 CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (*CPS1*) **negative**
 CARNITINE DEFICIENCY (*SLC22A5*) **negative**
 CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY (*CPT1A*) **negative**
 CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (*CPT2*) **negative**
 CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (*SLC25A20*) **negative**
 CARPENTER SYNDROME (*RAB23*) **negative**
 CARTILAGE-HAIR HYPOPLASIA (*RMRP*) **negative**
 CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (*CASQ2*) **negative**
 CD59-MEDIATED HEMOLYTIC ANEMIA (*CD59*) **negative**
 CEP152-RELATED MICROCEPHALY (*CEP152*) **negative**
 CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA (CEDNIK) SYNDROME (*SNAP29*) **negative**
 CEREBROTENDINOUS XANTHOMATOSIS (*CYP27A1*) **negative**
 CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE C (*PLEKHG5*) **negative**
 CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (*NDRG1*) **negative**
 CHEDIAK-HIGASHI SYNDROME (*LYST*) **negative**
 CHOREOACANTHOCYTOSIS (*VPS13A*) **negative**
 CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (*CYBA*) **negative**
 CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (*NCF2*) **negative**
 CILIOPATHIES, RPGRIP1L-RELATED (*RPGRIP1L*) **negative**
 CITRIN DEFICIENCY (*SLC25A13*) **negative**
 CITRULLINEMIA, TYPE 1 (*ASS1*) **negative**
 CLN10 DISEASE (*CTSD*) **negative**
 COHEN SYNDROME (*VPS13B*) **negative**
 COL11A2-RELATED CONDITIONS (*COL11A2*) **negative**
 COMBINED MALONIC AND METHYLMALONIC ACIDURIA (*ACSF3*) **negative**
 COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (*GFM1*) **negative**
 COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (*TFSM*) **negative**



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C

COMBINED PITUITARY HORMONE DEFICIENCY 1 (*POU1F1*) **negative**
 COMBINED PITUITARY HORMONE DEFICIENCY-2 (*PROP1*) **negative**
 CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (*CYP11B1*) **negative**
 CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (*CYP17A1*) **negative**
 CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (*CYP21A2*) **negative**
 CONGENITAL ADRENAL INSUFFICIENCY, *CYP11A1*-RELATED (*CYP11A1*) **negative**
 CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (*MPL*) **negative**
 CONGENITAL CHRONIC DIARRHEA (*DGAT1*) **negative**
 CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, *ALG1*-RELATED (*ALG1*) **negative**
 CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, *PMM2*-Related (*PMM2*) **negative**
 CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (*MPL*) **negative**
 CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (*ALG6*) **negative**
 CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (*SEC23B*) **negative**
 CONGENITAL FINNISH NEPHROSIS (*NPHS1*) **negative**
 CONGENITAL HYDROCEPHALUS 1 (*CCDC88C*) **negative**
 CONGENITAL HYPERINSULINISM, *KCNJ11*-Related (*KCNJ11*) **negative**
 CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (*CIPA*) (*NTRK1*) **negative**
 CONGENITAL MYASTHENIC SYNDROME, *CHAT*-RELATED (*CHAT*) **negative**
 CONGENITAL MYASTHENIC SYNDROME, *CHRNA*-RELATED (*CHRNA*) **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**
 COSTEFF SYNDROME (3-METHYLGLUTACONIC ACIDURIA, TYPE 3) (*OPA3*) **negative**
 CRB1-RELATED RETINAL DYSTROPHIES (*CRB1*) **negative**
 CYSTIC FIBROSIS (*CFTR*) **negative**
 CYSTINOSIS (*CTNS*) **negative**
 CYTOCHROME C OXIDASE DEFICIENCY, *PET100*-RELATED (*PET100*) **negative**
 CYTOCHROME P450 OXIDOREDUCTASE DEFICIENCY (*POR*) **negative**

D

D-BIFUNCTIONAL PROTEIN DEFICIENCY (*HSD17B4*) **negative**
 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, *RTKL*-RELATED (*RTKL*) **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/DESBQUOIS DYSPLASIA 1 (*CANT1*) **negative**
 ERCC6-RELATED DISORDERS (*ERCC6*) **negative**
 ERCC8-RELATED DISORDERS (*ERCC8*) **negative**
 ETHYLMALONIC ENCEPHALOPATHY (*ETHE1*) **negative**

F

FACTOR XI DEFICIENCY (*F11*) **negative**

FAMILIAL DYSAUTONOMIA (*IKBKAP*) **negative**
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, *PRF1*-RELATED (*PRF1*) **negative**
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, *STX11*-RELATED (*STX11*) **negative**
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, *STXBP2*-RELATED (*STXBP2*) **negative**
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, *UNC13D*-RELATED (*UNC13D*) **negative**
 FAMILIAL HYPERCHOLESTEROLEMIA, *LDLRAP1*-RELATED (*LDLRAP1*) **negative**
 FAMILIAL HYPERCHOLESTEROLEMIA, *LDLR*-RELATED (*LDLR*) **negative**
 FAMILIAL HYPERINSULINISM, *ABCC8*-RELATED (*ABCC8*) **negative**
 FAMILIAL NEPHROGENIC DIABETES INSIPIDUS, *AQP2*-RELATED (*AQP2*) **negative**
 FANCONI ANEMIA, GROUP A (*FANCA*) **negative**
 FANCONI ANEMIA, GROUP C (*FANCC*) **negative**
 FANCONI ANEMIA, GROUP D2 (*FANCD2*) **negative**
 FANCONI ANEMIA, GROUP E (*FANCE*) **negative**
 FANCONI ANEMIA, GROUP F (*FANCF*) **negative**
 FANCONI ANEMIA, GROUP G (*FANCG*) **negative**
 FANCONI ANEMIA, GROUP I (*FANCI*) **negative**
 FANCONI ANEMIA, GROUP J (*BRIP1*) **negative**
 FANCONI ANEMIA, GROUP L (*FANCL*) **negative**
 FARBER LIPOGRANULOMATOSIS (*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, *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**



Patient Information

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Test Information

Ordering Physician: [REDACTED]



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H

HEREDITARY FRUCTOSE INTOLERANCE (ALDOB) **negative**
 HEREDITARY HEMOCHROMATOSIS TYPE 2B (HAMP) **negative**
 HEREDITARY SPASTIC PARAPARESIS, TYPE 49 (TECP2) **negative**
 HEREDITARY SPASTIC PARAPLEGIA, CYP7B1-RELATED (CYP7B1) **negative**
 HERMANSKY-PUDLAK SYNDROME, AP3B1-RELATED (AP3B1) **negative**
 HERMANSKY-PUDLAK SYNDROME, BLOC153-RELATED (BLOC153) **negative**
 HERMANSKY-PUDLAK SYNDROME, BLOC156-RELATED (BLOC156) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED (HPS1) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS3-RELATED (HPS3) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS4-RELATED (HPS4) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS5-RELATED (HPS5) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS6-RELATED (HPS6) **negative**
 HOLOCARBOXYLASE SYNTHETASE DEFICIENCY (HLCS) **negative**
 HOMOCYSTINURIA AND MEGALOBlastic ANEMIA TYPE CBLG (MTR) **negative**
 HOMOCYSTINURIA DUE TO DEFICIENCY OF MTHFR (MTHFR) **negative**
 HOMOCYSTINURIA, CBS-RELATED (CBS) **negative**
 HOMOCYSTINURIA, Type cblE (MTRR) **negative**
 HYDROLETHALUS SYNDROME (HLYS1) **negative**
 HYPER-IGM IMMUNODEFICIENCY (CD40) **negative**
 HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA (HHH SYNDROME) (SLC25A15) **negative**
 HYPERPHOSPHATEMIC FAMILIAL TUMORAL CALCINOSIS, GALNT3-RELATED (GALNT3) **negative**
 HYPOMYELINATING LEUKODYSTROPHY 12 (VPS11) **negative**
 HYPOPHOSPHATASIA, ALPL-RELATED (ALPL) **negative**

I

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

J

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

KKRABBE DISEASE (GALC) **negative****L**

LAMELLAR ICHTHYOSIS, TYPE 1 (TGM1) **negative**
 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 (SGDG) **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**
 MECP-RELATED NEUROLOGIC DISORDER (MECP) **negative**
 MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (ACADM) **negative**
 MEDNIK SYNDROME (AP1S1) **negative**
 MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS (MLC1) **negative**
 MEROSIN-DEFICIENT MUSCULAR DYSTROPHY (LAMA2) **negative**
 METABOLIC ENCEPHALOPATHY AND ARRHYTHMIAS, TANGO2-RELATED (TANGO2) **negative**
 METACHROMATIC LEUKODYSTROPHY, ARSA-RELATED (ARSA) **negative**
 METACHROMATIC LEUKODYSTROPHY, PSAP-RELATED (PSAP) **negative**
 METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CBLF (LMBRD1) **negative**
 METHYLMALONIC ACIDEMIA, MCEE-RELATED (MCEE) **negative**
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CBLF (MMACHC) **negative**
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CblD (MMADHC) **negative**



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Test Information

Ordering Physician: [REDACTED]



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M

METHYLMALONIC ACIDURIA, MMAA-RELATED (MMAA) **negative**
METHYLMALONIC ACIDURIA, MMAB-RELATED (MMAB) **negative**
METHYLMALONIC ACIDURIA, TYPE MUT(0) (MUT) **negative**
MEVALONIC KINASE DEFICIENCY (MKV) **negative**
MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II (PCNT) **negative**
MICROPHTHALMIA / ANOPHTHALMIA, VSX2-RELATED (VSX2) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, ACAD9-RELATED (ACAD9) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NDUFAF5-RELATED (NDUFAF5) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NDUFS6-RELATED (NDUFS6) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 1 (NDUFS4) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 10 (NDUFAF2) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 17 (NDUFAF6) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 19 (FOXRED1) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 3 (NDUFS7) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 4 (NDUFV1) **negative**
MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 2, SCO2-RELATED (SCO2) **negative**
MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 6 (COX15) **negative**
MITOCHONDRIAL DNA DEPLETION SYNDROME 2 (TK2) **negative**
MITOCHONDRIAL DNA DEPLETION SYNDROME 3 (DGUOK) **negative**
MITOCHONDRIAL MYOPATHY AND SIDEROBLASTIC ANEMIA (MLASA) (PUS1) **negative**
MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (HADHB) **negative**
MOLYBDENUM COFACTOR DEFICIENCY TYPE B (MOC52) **negative**
MOLYBDENUM COFACTOR DEFICIENCY, TYPE A (MOC51) **negative**
MUCOLIPIDOSIS II/III A (GNPTAB) **negative**
MUCOLIPIDOSIS III GAMMA (GNPTG) **negative**
MUCOLIPIDOSIS, TYPE IV (MCOLN1) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE I (HURLER SYNDROME) (IDUA) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III A (SANFILIPPO A) (SGSH) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III B (SANFILIPPO B) (NAGLU) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III C (SANFILIPPO C) (HGSNAT) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III D (SANFILIPPO D) (GNS) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE IV A (MORQUIO SYNDROME) (GALNS) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE IV B/GM1 GANGLIOSIDOSIS (GLB1) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE IX (HYAL1) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE VI (MAROTEAUX-LAMY) (ARSB) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE VII (GUSB) **negative**
MULIBREY NANISM (TRIM37) **negative**
MULTIPLE PTERYGIUM SYNDROME, CHRNG-RELATED/ESCOBAR SYNDROME (CHRNA) **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-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, PJVK-RELATED (PJVK) **negative**
NONSYNDROMIC HEARING LOSS, SYNE4-RELATED (SYNE4) **negative**
NONSYNDROMIC HEARING LOSS, TMC1-RELATED (TMC1) **negative**
NONSYNDROMIC HEARING LOSS, TMPS53-RELATED (TMPS53) **negative**
NONSYNDROMIC INTELLECTUAL DISABILITY (CC2D1A) **negative**
NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (SAMD9) **negative**

O

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

P

PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION (PANK2) **negative**
PAPILLON 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) **see first page**
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 (SEPS2) **negative**
PONTOCEREBELLAR HYPOPLASIA, VPS53-RELATED (VPS53) **negative**
PRIMARY CILIARY DYSKINESIA, CCDC103-RELATED (CCDC103) **negative**
PRIMARY CILIARY DYSKINESIA, CCDC39-RELATED (CCDC39) **negative**
PRIMARY CILIARY DYSKINESIA, DNAH11-RELATED (DNAH11) **negative**
PRIMARY CILIARY DYSKINESIA, DNAH5-RELATED (DNAH5) **negative**
PRIMARY CILIARY DYSKINESIA, DNAI1-RELATED (DNAI1) **negative**
PRIMARY CILIARY DYSKINESIA, DNAI2-RELATED (DNAI2) **negative**
PRIMARY CONGENITAL GLAUCOMA/PETERS ANOMALY (CYP1B1) **negative**
PRIMARY HYPEROXALURIA, TYPE 1 (AGXT) **negative**
PRIMARY HYPEROXALURIA, TYPE 2 (GRHPR) **negative**
PRIMARY HYPEROXALURIA, TYPE 3 (HOGA1) **negative**
PRIMARY MICROCEPHALY 1, AUTOSOMAL RECESSIVE (MCPH1) **negative**
PROGRESSIVE EARLY-ONSET ENCEPHALOPATHY WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM (TBCD) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, ABCB4-RELATED (ABCB4) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 1 (PFIC1) (ATP8B1) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 2 (ABCB11) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 4 (PFIC4) (TJP2) **negative**
PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA (CCN6) **negative**
PROLIDASE DEFICIENCY (PEPD) **negative**
PROPIONIC ACIDEMIA, PCCA-RELATED (PCCA) **negative**
PROPIONIC ACIDEMIA, PCCB-RELATED (PCCB) **negative**
PSEUDOXANTHOMA ELASTICUM (ABCC6) **negative**



Patient Information

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Test Information

Ordering Physician: [REDACTED]



Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

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P

PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (PCD) DEFICIENCY (*PCBD1*) **negative**
 PYCNODYSOSTOSIS (*CTSK*) **negative**
 PYRIDOXAL 5'-PHOSPHATE-DEPENDENT EPILEPSY (*PNPO*) **negative**
 PYRIDOXINE-DEPENDENT EPILEPSY (*ALDH7A1*) **negative**
 PYRUVATE CARBOXYLASE DEFICIENCY (*PC*) **negative**
 PYRUVATE DEHYDROGENASE DEFICIENCY, PDHB-RELATED (*PDHB*) **negative**

R

REFSUM DISEASE, PHYH-RELATED (*PHYH*) **negative**
 RENAL TUBULAR ACIDOSIS AND DEAFNESS, ATP6V1B1-RELATED (*ATP6V1B1*) **negative**
 RENAL TUBULAR ACIDOSIS, PROXIMAL, WITH OCULAR ABNORMALITIES AND MENTAL RETARDATION (*SLC4A4*) **negative**
 RETINITIS PIGMENTOSA 25 (*EYS*) **negative**
 RETINITIS PIGMENTOSA 26 (*CERKL*) **negative**
 RETINITIS PIGMENTOSA 28 (*FAM161A*) **negative**
 RETINITIS PIGMENTOSA 36 (*PRCD*) **negative**
 RETINITIS PIGMENTOSA 59 (*DHDDS*) **negative**
 RETINITIS PIGMENTOSA 62 (*MAK*) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1 (*PEX7*) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2 (*GNPAT*) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3 (*AGPS*) **negative**
 RLB1-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 (*SMARCA1*) **negative**
 SCHINDLER DISEASE (*NAGA*) **negative**
 SEGAWA SYNDROME, TH-RELATED (*TH*) **negative**
 SENIOR-LOKEN SYNDROME 4/NEPHRONOPHTHISIS 4 (*NPHP4*) **negative**
 SEPIAPTERIN REDUCTASE DEFICIENCY (*SPR*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3D-RELATED (*CD3D*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3E-RELATED (*CD3E*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), 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 (*DYNC2H1*) **negative**
 SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED (*SBDS*) **negative**
 SIALIDOSIS (*NEU1*) **negative**
 SJOGREN-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 (*TCN2*) **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**
 VLDL-ASSOCIATED CEREBELLAR HYPOPLASIA (*VLDLR*) **negative**

W

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

X

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

Z

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



Patient Information

Patient Name: Donor 8416

Test Information

Ordering Physician: [REDACTED]



Horizon™
Advanced carrier screening

Date Of Birth: [REDACTED]

Clinic Information: Fairfax Cryobank

Case File ID: [REDACTED]

Report Date: 03/04/2026

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



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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-CRYL1 critical region 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. Sequencing and CNV analysis may have reduced sensitivity due to high sequence homology.

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.



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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

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.

