

SPERM DONOR GENETIC TESTING SUMMARY

Donor # 7583

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

Last Updated: 12/22/2025

Donor Reported Ancestry: Kyrgyz, Russian

Jewish Ancestry: No

Genetic Test*	Result	Comments Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, and other hemoglobinopathies
Expanded Genetic Disease Carrier Screening Panel attached - 549 diseases by gene sequencing and del/dup analysis.	Carrier: Non-Syndromic Hearing Loss, GJB2-Related (GJB2) Negative for other genes tested	Partner testing is recommended before using this donor

*No single test can screen for all genetic disorders. A negative screening result significantly reduces, but cannot eliminate, the risk for these conditions in a pregnancy.

**Donor residual risk is the chance the donor is still a carrier after testing negative.

Patient Information	
Patient Name:	Donor 7583
Date Of Birth:	[REDACTED]
Gender:	Male
Ethnicity:	Northern European Caucasian
Patient ID:	N/A
Medical Record #:	[REDACTED]
Collection Kit:	[REDACTED]
Accession ID:	N/A
Case File ID:	[REDACTED]

Test Information	
Ordering Physician:	[REDACTED]
Clinic Information:	Fairfax Cryobank
Phone:	[REDACTED]
Report Date:	03/28/2025
Sample Collected:	03/17/2025
Sample Received:	03/18/2025
Sample Type:	Blood



CARRIER SCREENING REPORT

ABOUT THIS SCREEN: Horizon™ is a carrier screen for specific autosomal recessive and X-linked diseases. This information can help patients learn their risk of having a child with specific genetic conditions.

ORDER SELECTED: The Horizon Custom panel was ordered for this patient. Males are not screened for X-linked diseases

FINAL RESULTS SUMMARY:



CARRIER for Non-Syndromic Hearing Loss, GJB2-Related

Positive for the pathogenic variant c.235del (p.L79Cfs*3) in the GJB2 gene. Although most variants in this gene are associated with an autosomal recessive form of NON-SYNDROMIC HEARING LOSS, GJB2-RELATED, some rare GJB2 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 NON-SYNDROMIC HEARING LOSS, GJB2-RELATED, 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

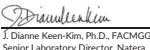
No other pathogenic variants were detected in the genes that were screened. The patient's remaining carrier risk after the negative screening results is listed for each disease/gene on the Horizon website at <https://www.natera.com/panel-option/h-all/>. Please see the following pages of this report for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

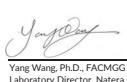
RECOMMENDATIONS

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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NON-SYNDROMIC HEARING LOSS, GJB2-RELATED

Understanding Your Horizon Carrier Screen Results

What is Non-Syndromic Hearing Loss, GJB2-Related?

Non-Syndromic Hearing Loss, GJB2-Related (also called DFNB1) is an inherited disorder that causes early-onset hearing loss. "Non-syndromic" means that no other parts of the body are affected, making hearing loss the only symptom of this condition. In Non-Syndromic Hearing Loss, GJB2-Related, hearing loss is typically present at birth (congenital). However, some children have normal hearing at birth and develop hearing loss during childhood. The severity varies from mild to profound sensorineural hearing loss. The treatment for hearing loss includes hearing aids and, in some cases, cochlear implants. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov). Non-Syndromic Hearing Loss, GJB2-Related does not cause other health problems.

What causes Non-Syndromic Hearing Loss, GJB2-Related?

Non-Syndromic Hearing Loss, GJB2-Related is caused by a gene change, or mutation, in both copies of the GJB2 gene pair (also known as DFNB1). These mutations cause the genes to not work properly or not work at all. The function of the GJB2 genes is to make a protein that is important for hearing. When both copies of the GJB2 gene do not work correctly, it leads to Non-Syndromic Hearing Loss, GJB2-Related. Non-Syndromic Hearing Loss, GJB2-Related is inherited in an autosomal recessive manner. This means that, in most cases, both parents must be carriers of a mutation in one copy of the GJB2 gene to have a child with Non-Syndromic Hearing Loss, GJB2-Related. People who are carriers for Non-Syndromic Hearing Loss, GJB2-Related are usually healthy and usually do not have Non-Syndromic Hearing Loss 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 Non-Syndromic Hearing Loss, GJB2-Related, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their GJB2 gene mutations to the child, who will then have Non-Syndromic Hearing Loss, GJB2-Related. Very rarely, carriers of a single GJB2 mutation will have inherited hearing loss with or without other symptoms. These individuals usually have one parent who is also affected. This type of inheritance, where having only one mutation causes symptoms, is called autosomal dominant. When a person with autosomal dominant hearing loss has a child, there is a 50%, or 1 in 2, chance with each pregnancy of having a child who will also develop this type of hearing loss. It is sometimes, but not always, possible to determine whether a specific mutation in the GJB2 gene will cause autosomal recessive Non-Syndromic Hearing Loss or an autosomal dominant type of hearing loss. Individuals found to carry more than one mutation for Non-Syndromic Hearing Loss, GJB2-Related should discuss their risk for having an affected child and any potential effects to their own hearing with their health care provider.

What can I do next?

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website (www.nsgc.org). You may wish to share your carrier screening results with your health care providers, especially if you have a family history of hearing loss or have concerns about your own hearing. 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 Non-Syndromic Hearing Loss, GJB2-Related ordered by a health care professional. If your partner is not found to be a carrier for Non-Syndromic Hearing Loss, GJB2-Related, your risk of having a child with Non-Syndromic Hearing Loss, GJB2-Related is greatly reduced. Couples at risk of having a baby with Non-Syndromic Hearing Loss, GJB2-Related can opt to have prenatal diagnosis done through chorionic villus sampling (CVS) or amniocentesis during pregnancy or can choose to test the baby after birth for this condition. If you are not yet pregnant, your partner can have carrier screening for Non-Syndromic Hearing Loss, GJB2-Related ordered by a health care professional. If your partner is found to be a carrier for Non-Syndromic Hearing Loss, GJB2-Related, you have several reproductive options to consider:

- Natural pregnancy with or without prenatal diagnosis of the fetus or testing the baby after birth for Non-Syndromic Hearing Loss, GJB2-Related
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for Non-Syndromic Hearing Loss, GJB2-Related
- Adoption or use of a sperm or egg donor who is not a carrier for Non-Syndromic Hearing Loss, GJB2-Related

What resources are available?

- Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/nonsyndromic-hearing-loss>
- Prenatal diagnosis done through CVS: <http://www.marchofdimes.org/chorionic-villus-sampling.aspx>
- Prenatal diagnosis done through Amniocentesis: <http://www.marchofdimes.org/amniocentesis.aspx>
- Preimplantation genetic diagnosis (PGD) with IVF: <http://www.natera.com/spectrum>

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Clinic Information: Fairfax Cryobank

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

Report Date: 03/28/2025



VARIANT DETAILS

GJB2, c.235del (p.L79Cfs*3), pathogenic

- The c.235del (p.L79Cfs*3) variant in the GJB2 gene has been observed at a frequency of 0.0474% 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 nonsyndromic hearing loss and deafness (DFNB) 1 (PMID: 10633133).
- This premature termination variant is predicted to cause nonsense-mediated decay (NMD) in a gene where loss-of-function is a known mechanism of disease.
- This variant has been reported in ClinVar [ID: 17014].

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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

5
5-ALPHA-REDUCTASE DEFICIENCY (SRD5A2) negative

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

A
ABCA4-RELATED CONDITIONS (ABCA4) negative
ABETALIPOPROTEINEMIA (MTP) negative
ACHONDROGENESIS, TYPE 1B (SLC2A2) negative
ACHROMATOPSY, CNGB3-RELATED (CNGB3) negative
ACRODERMATITIS ENTEROPATHICA (SLC39A4) negative
ACTION MYOCLONUS-RENAL FAILURE (AMRF) SYNDROME (SCARB2) negative
ACUTE INFANTILE LIVER FAILURE, TRMU-RELATED (TRMU) negative
ACYL-COA OXIDASE I DEFICIENCY (ACOX1) negative
AICARDI-GOUTIÈRES SYNDROME (SAMHD1) negative
AICARDI-GOUTIÈRES SYNDROME, RNASEH2A-RELATED (RNASEH2A) negative
AICARDI-GOUTIÈRES SYNDROME, RNASEH2B-RELATED (RNASEH2B) negative
AICARDI-GOUTIÈRES SYNDROME, RNASEH2C-RELATED (RNASEH2C) negative
AICARDI-GOUTIÈRES SYNDROME, TREX1-RELATED (TREX1) negative
ALPHA-MANNOSEIDOSIS (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 (TPPA) negative
ATAxia-TELangiectasia (ATM) negative
ATAxia-TELangiectasia-LIKE DISORDER 1 (MRE11) negative
ATRANSFERRINEMIA (TF) negative
AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (SLC35A3) negative
AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (AIRE) negative
AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (ARCI), SLC27A4-RELATED (SLC27A4) negative
AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (SACS) negative

B
BARDET-BIEDL SYNDROME, ARL6-RELATED (ARL6) negative
BARDET-BIEDL SYNDROME, BBS10-RELATED (BBS10) negative
BARDET-BIEDL SYNDROME, BBS12-RELATED (BBS12) negative
BARDET-BIEDL SYNDROME, BBS1-RELATED (BBS1) negative
BARDET-BIEDL SYNDROME, BBS2-RELATED (BBS2) negative
BARDET-BIEDL SYNDROME, BBS4-RELATED (BBS4) negative
BARDET-BIEDL SYNDROME, BBS5-RELATED (BBS5) negative
BARDET-BIEDL SYNDROME, BBS7-RELATED (BBS7) negative
BARDET-BIEDL SYNDROME, BBS9-RELATED (BBS9) negative
BARDET-BIEDL SYNDROME, TTC8-RELATED (TTC8) negative
BARE LYMPHOCYTE SYNDROME, CITA-RELATED (CITA) negative
BARTTER SYNDROME, BSND-RELATED (BSND) negative
BARTTER SYNDROME, KCNJ1-RELATED (KCNJ1) negative
BARTTER SYNDROME, SLC12A1-RELATED (SLC12A1) negative
BATTEN DISEASE, CLN3-RELATED (CLN3) negative
BETA-HEMOGLOBINOPATHIES (HBB) negative
BETA-KETOThIOLASE DEFICIENCY (ACAT1) negative
BETA-MANNOSEIDOSIS (MANBA) negative
BETA-UREIDOPROPIONASE DEFICIENCY (UPB1) negative
BILATERAL FRONTOPIRIETAL POLYMICROGYRIA (GPR56) negative

C
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

D
CARNAVAN 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
CEREBROENDINOS 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, RGPRI1-RELATED (RGPRI1) 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 (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, CHRNE-RELATED (CHRNE) negative
CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED (COLQ) negative
CONGENITAL MYASTHENIC SYNDROME, DOK7-RELATED (DOK7) negative
CONGENITAL MYASTHENIC SYNDROME, RAPSN-RELATED (RAPSN) negative
CONGENITAL NEPHROPATHY, 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

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D

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

E

EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY, CAD-RELATED (CAD) negative
 EHlers-DANLOS SYNDROME TYPE VI (PLOD1) negative
 EHlers-DANLOS SYNDROME, CLASSIC-LIKE, TNXB-RELATED (TNXB) negative
 EHlers-DANLOS SYNDROME, TYPE VII C (ADAMTS2) negative
 ELLIS-VAN CREVELD SYNDROME, EVC2-RELATED (EVC2) negative
 ELLIS-VAN CREVELD SYNDROME, EVC-RELATED (EVC) negative
 ENHANCED S-CONE SYNDROME (NR2E3) negative
 EPIMERASE DEFICIENCY (GALACTOSEMIA TYPE III) (GALE) negative
 EPIPHYSEAL DYSPLASIA, MULTIPLE, 7/DESBUVOIS 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 LIPOGANULOMATOSIS (ASAHI1) negative
 FOVEAL HYPOMELANOSIS (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, FUC4-RELATED (FUC4) negative
 FUMARASE DEFICIENCY (FH) negative

G

GABA-TRANSAMINASE DEFICIENCY (ABAT) negative
 GALACTOKINASE DEFICIENCY (GALACTOSEMIA, TYPE II) (GALK1) negative
 GALACTOSEMIA (GALT) negative
 GALACTOSIALIDOSIS (CTSA) negative
 GAUCHER DISEASE (GBA) negative
 GCH1-RELATED CONDITIONS (GCH1) negative
 GDF5-RELATED CONDITIONS (GDF5) negative
 GERODERMA OSTEODYPLASTICA (GORAB) negative
 GITELMAN SYNDROME (SLC12A3) negative
 GLANZMANN THROMBASTHENIA (ITGB3) negative
 GLUTARIC ACIDEMIA, TYPE 1 (GCDH) negative
 GLUTARIC ACIDEMIA, TYPE 2A (ETFA) negative
 GLUTARIC ACIDEMIA, TYPE 2B (ETFB) negative
 GLUTARIC ACIDEMIA, TYPE 2C (ETFDH) negative
 GLUTATHIONE SYNTHETASE DEFICIENCY (GSS) negative
 GLYCINE ENCEPHALOPATHY, AMT-RELATED (AMT) negative
 GLYCINE ENCEPHALOPATHY, GLDC-RELATED (GLDC) negative
 GLYCOGEN STORAGE DISEASE TYPE 5 (McArdle Disease) (PYGM) negative
 GLYCOGEN STORAGE DISEASE TYPE IXB (PHKB) negative
 GLYCOGEN STORAGE DISEASE TYPE IXC (PHKG2) negative
 GLYCOGEN STORAGE DISEASE, TYPE 1a (G6PC) negative
 GLYCOGEN STORAGE DISEASE, TYPE 1b (SLC37A4) negative
 GLYCOGEN STORAGE DISEASE, TYPE 2 (POMPE DISEASE) (GAA) negative
 GLYCOGEN STORAGE DISEASE, TYPE 3 (AGL) negative
 GLYCOGEN STORAGE DISEASE, TYPE 4 (GBE1) negative
 GLYCOGEN STORAGE DISEASE, TYPE 7 (PFKM) negative

Test Information

Ordering Physician: [REDACTED]

Clinic Information: Fairfax Cryobank



GRACILE SYNDROME (BCS1L) negative
 GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY (GAMT) negative

H

HARLEQUIN ICHTHYOSIS (ABCA12) negative
 HEME OXYGENASE 1 DEFICIENCY (HMOX1) negative
 HEMOCHROMATOSIS TYPE 2A (HFE2) negative
 HEMOCHROMATOSIS, TYPE 3, TFR2-Related (TFR2) negative
 HEPATOCEREBRAL MITOCHONDRIAL DNA DEPLETION SYNDROME, MPV17-RELATED (MPV17) negative
 HEREDITARY FRUCTOSE INTOLERANCE (ALDOB) negative
 HEREDITARY HEMOCHROMATOSIS TYPE 2B (HAMP) negative
 HEREDITARY SPASTIC PARAPARESIS, TYPE 49 (TECPR2) negative
 HEREDITARY SPASTIC PARAPLEGIA, CYP7B1-RELATED (CYP7B1) negative
 HERMANSKY-PUDLAK SYNDROME, AP3B1-RELATED (AP3B1) negative
 HERMANSKY-PUDLAK SYNDROME, BLOC1S3-RELATED (BLOC1S3) negative
 HERMANSKY-PUDLAK SYNDROME, BLOC1S6-RELATED (BLOC1S6) negative
 HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED (HPS1) negative
 HERMANSKY-PUDLAK SYNDROME, HPS3-RELATED (HPS3) negative
 HERMANSKY-PUDLAK SYNDROME, HPS4-RELATED (HPS4) negative
 HERMANSKY-PUDLAK SYNDROME, HPS5-RELATED (HPS5) negative
 HERMANSKY-PUDLAK SYNDROME, HPS6-RELATED (HPS6) negative
 HOLOCARBOXYLASE SYNTHETASE DEFICIENCY (HLCS) negative
 HOMOCYSTINURIA AND MEGALOBLASTIC ANEMIA TYPE CBLG (MTR) negative
 HOMOCYSTINURIA DUE TO DEFICIENCY OF MTHFR (MTHFR) negative
 HOMOCYSTINURIA, CBS-RELATED (CBS) negative
 HOMOCYSTINURIA, Type cblE (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 (INVS) 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, CPLAN1-RELATED / OROFACIODIGITAL SYNDROME 6 (CPLAN1) 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

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Test Information

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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 2G (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 (MKKS1) negative
 MECR-RELATED NEUROLOGIC DISORDER (MECR) negative
 MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (ACADM) negative
 MEDNIK SYNDROME (AP1S1) negative
 MEGAENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS (MLC1) negative
 MEROSIN-DEFICIENT MUSCULAR DYSTROPHY (LAMA2) negative
 METABOLIC ENCEPHALOPATHY AND ARRHYTHMIAS, TANGO2-RELATED (TANGO2) negative
 METACHROMATIC LEUKODYSTROPHY, ARSA-RELATED (ARSA) negative
 METACHROMATIC LEUKODYSTROPHY, PSAP-RELATED (PSAP) negative
 METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CBLF (LMBRD1) negative
 METHYLMALONIC ACIDEMIA, MCEE-RELATED (MCEE) negative
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CBLC (MMACHC) negative
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE Cb1D (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
 MICROPTHALMIA / 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 1 DEFICIENCY, NUCLEAR TYPE 1 (NDUFS4) negative
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NUCLEAR TYPE 10 (NDUFAF2) negative
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NUCLEAR TYPE 17 (NDUFAF6) negative
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NUCLEAR TYPE 19 (FOXRED1) negative
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NUCLEAR TYPE 3 (NDUFS7) negative
 MITOCHONDRIAL COMPLEX 1 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

Test Information

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MITOCHONDRIAL DNA DEPLETION SYNDROME 3 (DGUOK) negative
 MITOCHONDRIAL MYOPATHY AND SIDEROBLOMATIC ANEMIA (MLASA1) (PUS1) negative
 MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (HADHB) negative
 MOLYBDENUM COFACTOR DEFICIENCY TYPE B (MOCSD2) negative
 MOLYBDENUM COFACTOR DEFICIENCY, TYPE A (MOCSD1) 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 (CHRNG) negative
 MULTIPLE SULFATASE DEFICIENCY (SUMF1) negative
 MUSCLE-EYE-BRAIN DISEASE, POMGNT1-RELATED (POMGNT1) negative
 MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (RYXLT1) 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) see first page
 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 (C2D1A) negative
 NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (SAMD9) negative

O

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

P

PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION (PANK2) negative
 PAPILLON LEFÈVRE SYNDROME (CTSC) negative
 PARKINSON DISEASE 15 (FBXO7) negative
 PENDRED SYNDROME (SLC26A4) negative
 PERLMAN SYNDROME (DIS3L2) negative
 PGM3-CONGENITAL DISORDER OF GLYCOSYLATION (PGM3) negative
 PHENYLKETONURIA (PAH) negative
 PIGN-CONGENITAL DISORDER OF GLYCOSYLATION (PIGN) negative
 PITUITARY HORMONE DEFICIENCY, COMBINED 3 (LHX3) negative
 POLG-RELATED DISORDERS (POLG) negative

Patient Information

Patient Name: Donor 7583

Test Information

Ordering Physician: [REDACTED]

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03/28/2025

P
 POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE (PKHD1) negative
 PONTOCEREBELLAR HYPOPLASIA, EXOSC3-RELATED (EXOSC3) negative
 PONTOCEREBELLAR HYPOPLASIA, RARS2-RELATED (RARS2) negative
 PONTOCEREBELLAR HYPOPLASIA, TSEN2-RELATED (TSEN2) negative
 PONTOCEREBELLAR HYPOPLASIA, TSEN54-RELATED (TSEN54) negative
 PONTOCEREBELLAR HYPOPLASIA, TYPE 1A (VRK1) negative
 PONTOCEREBELLAR HYPOPLASIA, TYPE 2D (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, 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) (ATP88B1) negative
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 2 (ABC811) 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
 PYCNOIDSOSTOSIS (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
 RLPB1-RELATED RETINOPATHY (RLPB1) 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, 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
 SJÖGREN-LARSSON SYNDROME (ALDH3A2) negative
 SMITH-LEMLI-OPITZ SYNDROME (DHCR7) negative
 SPASTIC PARAPLEGIA, TYPE 15 (ZFYVE26) negative

T
 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

V
 TAY-SACHS DISEASE (HEXA) negative
 TBCE-RELATED CONDITIONS (TBCE) negative
 THIAMINE-RESPONSIVE MEALOBLASTIC 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
 TRIMETHYLAURINURIA (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

W
 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

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 7583

Test Information

Ordering Physician: [REDACTED]

Date Of Birth: [REDACTED]

Clinic Information: Fairfax Cryobank

Case File ID: [REDACTED]

Report Date: 03/28/2025

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-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

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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.