Fairfax Cryobank

3015 Williams Drive, Fairfax, VA 22031
info@fairfaxcryobank.com ● www.fairfaxcryobank.com
(800)338-8407 phone
(703)698-3933 fax

SPERM DONOR GENETIC TESTING SUMMARY

Donor # 8107

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

Last Updated: 11/11/2025

Donor Reported Ancestry: Colombian

Jewish Ancestry: No

Genetic Test*	**Result**	**Comments** **Donor's Residual Risk****
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Expanded Genetic Disease Carrier Screening Panel attached - 549 diseases by gene sequencing and del/dup analysis.	**Carrier: Lamellar Ichthyosis, Type 1 (TGM1)** **Carrier: Thyroid Dyshormonogenesis 6 (DUOX2)** Negative for other genes tested.	Partner testing is recommended before using this donor. Most people with a variant in the DUOX2 gene are carriers of TDH6, but do not have the condition. Some people with a variant in this gene have symptoms of TDH6 as babies that go away as they age. Please see results for further information. Genetic counseling can be considered.

*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.

QuestQuanum

8107,DONOR ⚠

[redacted]

Collected: 05/06/2025 14:31
Received: 05/07/2025 14:13
Reported: 05/14/2025 17:56

[redacted]

⚠ Hemoglobinopathy Evaluation (FINAL)

Lab: AMD

Analyte	Value		
Hemoglobinopathy Evaluation			FINAL
Red Blood Cell Count	**5.58**	Reference Range: 4.20-5.80 Mill/uL	FINAL
⚠ HEMOGLOBIN	**17.4 H**	Reference Range: 13.2-17.1 g/dL	FINAL
Hematocrit			FINAL
⚠ Hematocrit	**52.9 H**	Reference Range: 38.5-50.0 %	FINAL
MCV	**94.8**	Reference Range: 80.0-100.0 fL	FINAL
MCH	**31.2**	Reference Range: 27.0-33.0 pg	FINAL
RDW	**12.0**	Reference Range: 11.0-15.0 %	FINAL
Hemoglobinopathy Evaluation			FINAL
Hemoglobin A	**97.4**	Reference Range: >96.0 %	FINAL
Hemoglobin F	**0.0**	Reference Range: <2.0 %	FINAL
Hemoglobin A2 (Quant)	**2.6**	Reference Range: 2.0-3.2 %	FINAL
Interpretation			FINAL

NORMAL PHENOTYPE

There are no variant peaks identified. The patient's hemogram shows elevated hemoglobin/hematocrit. It should be pointed out that elevated hemoglobin/hematocrit can be caused by a variety of conditions including high oxygen affinity hemoglobins which may not be detected by high-performance liquid chromatography (HPLC) and/or capillary zone electrophoresis (CZE). Rare variants hemoglobins have no separation from hemoglobin A by capillary zone electrophoresis (CZE) or high-performance liquid chromatography (HPLC).
If clinically indicated, Thalassemia and Hemoglobinopathy Comprehensive (TC 17365) should be considered.

[redacted]

[redacted]

Chromosome Analysis, Blood (FINAL)

Lab: AMD

Analyte	Value

Chromosome Analysis, Blood (FINAL)

Order ID: [redacted]

Specimen Type: Blood

Clinical Indication: Donor screening

RESULT:
NORMAL MALE KARYOTYPE

INTERPRETATION:
Chromosome analysis revealed normal G-band patterns within the limits of standard cytogenetic analysis.

Please expect the results of any other concurrent study in a separate report.

NOMENCLATURE:
46,XY

ASSAY INFORMATION:

Method:	G-Band (Digital Analysis: MetaSystems/Ikaros)
Cells Counted:	20
Band Level:	550
Cells Analyzed:	5
Cells Karyotyped:	3

This test does not address genetic disorders that cannot be detected by standard cytogenetic methods or rare events such as low level mosaicism or subtle rearrangements.
A portion of the testing was performed at AMD4.

Navnit S. Mitter, Ph.D., FACMG, Technical Director, Cytogenetics and Genomics, 703-802-7156, [AMD4]

Electronic Signature: 5/14/2025 5:12 PM

For additional information, please refer to
http://education.questdiagnostics.com/faq/chromsblood
(This link is being provided for informational/
educational purposes only).

Performing Sites

AMD Quest Diagnostics Nichols Institute, 14225 Newbrook Drive, Chantilly, VA 20151 Laboratory Director: Patrick W Mason, MD PhD

Key

Priority Out of Range ▲ Out of Range (PEND) Pending Result (PRE) Preliminary Result (FINAL) Final Result (RE) Reissued Result

Patient Information		Test Information	
Patient Name:	Donor 8107	Ordering Physician:	[redacted]
		Clinic Information:	Fairfax Cryobank
Date Of Birth:	[redacted]		
Gender:	Male		
Ethnicity:	Hispanic/Latin American	Phone:	
Patient ID:	N/A	Report Date:	05/20/2025
Medical Record #:	N/A	Sample Collected:	05/06/2025
Collection Kit:	[redacted]	Sample Received:	05/07/2025
Accession ID:	N/A	Sample Type:	Blood
Case File ID:	[redacted]		

horizon™ natera carrier screen

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 Lamellar Ichthyosis, Type 1

Positive for the likely pathogenic variant c.376C>T (p.R126C) in the TGM1 gene. If this individual's partner is a carrier for LAMELLAR ICHTHYOSIS, TYPE 1, their chance to have a child with this condition may be as high as 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

CARRIER for Thyroid Dyshormonogenesis 6

Positive for the likely pathogenic variant c.457C>T (p.Q153*) in the DUOX2 gene. If this individual's partner is a carrier for THYROID DYSHORMONOGENESIS 6, their chance to have a child with this condition may be as high as 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

Negative for 547 out of 549 diseases

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

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

RECOMMENDATIONS

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

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

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

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

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

natera® Conceive. Deliver.

Patient Information
Patient Name: Donor 8107

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

Test Information
Ordering Physician: [redacted]

Clinic Information: Fairfax Cryobank

Report Date: 05/20/2025

horizon
natera carrier screen

LAMELLAR ICHTHYOSIS, TYPE 1

Understanding Your Horizon Carrier Screen Results

What is Lamellar Ichthyosis, Type 1?

Lamellar Ichthyosis, Type 1 is an inherited disorder that affects the skin. Children with this disorder are born with a clear fitted covering over their body called a collodion membrane. After about 10 to 14 days of life this membrane peels off, leaving skin that is covered with white or brownish scales. Eyelids and lips may turn outward, fingernails and toenails may develop abnormally and hair loss may occur. Affected infants may develop skin infections, dehydration, and breathing problems, and in some cases, serious joint problems. Some infants will have improvement of their skin condition with time, and for others the skin problems may be lifelong. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov). Rarely, a specific mutation in the same gene causes a different type of ichthyosis, either 'Self-Improving Collodion Ichthyosis' in which affected babies are born with a collodion membrane but typically do not have severe lifelong skin problems, or 'Bathing Suit Ichthyosis' in which dark gray skin scales affect only the trunk area and not the limbs or face. It is sometimes, but not always possible to determine whether a specific mutation in the TGM1 gene will cause Lamellar Ichthyosis, Type 1 or one of these other milder forms of ichthyosis. The information below is about Lamellar Ichthyosis, Type 1, the most common disorder caused by mutations in the TGM1 gene. However, the other rare forms of Ichthyosis described above are inherited in the same manner as Lamellar Ichthyosis, Type 1 and have the same reproductive options.

What causes Lamellar Ichthyosis, Type 1?

Lamellar Ichthyosis, Type 1 is caused by a gene change, or mutation, in both copies of the TGM1 gene pair. These mutations cause the genes to not work properly or not work at all. The normal function of the TGM1 genes is to help in the development of the skin. When both copies of this gene do not work correctly it leads to the symptoms described above. Lamellar Ichthyosis, Type 1 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 TGM1 gene to have a child with Lamellar Ichthyosis, Type 1. People who are carriers for Lamellar Ichthyosis, Type 1 are usually healthy and do not have symptoms of Lamellar Ichthyosis, Type 1 nor do they have the disorder themselves. Usually a child inherits two copies of each gene, one copy from the mother and one copy from the father. If the mother and father are both carriers for Lamellar Ichthyosis, Type 1, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their TGM1 gene mutations to the child, who will then have this condition. Individuals found to carry more than one mutation for Lamellar Ichthyosis, Type 1 should discuss their risk for having an affected child with their health care provider. There are a number of other forms of Ichthyosis that are each caused by mutations in different genes. People who are carriers for Lamellar Ichthyosis, Type 1 are not likely to be at increased risk for having children with these other forms.

What can I do next?

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website (www.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 Lamellar Ichthyosis, Type 1 ordered by a health care professional. If your partner is not found to be a carrier for Lamellar Ichthyosis, Type 1, your risk of having a child with Lamellar Ichthyosis, Type 1 is greatly reduced. Couples at risk of having a baby with Lamellar Ichthyosis, Type 1 can opt to have prenatal diagnosis done through chorionic villus sampling (CVS) or amniocentesis during pregnancy or can choose to have the baby tested after birth for this condition. If you are not yet pregnant, your partner can have carrier screening for Lamellar Ichthyosis, Type 1 ordered by a health care professional. If your partner is found to be a carrier for Lamellar Ichthyosis, Type 1, you have several reproductive options to consider:

- Natural pregnancy with or without prenatal diagnosis of the fetus or testing the baby after birth for Lamellar Ichthyosis, Type 1
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for Lamellar Ichthyosis, Type 1
- Adoption or use of a sperm or egg donor who is not a carrier for Lamellar Ichthyosis, Type 1

What resources are available?

- Genetics Home Reference: http://ghr.nlm.nih.gov/condition/lamellar-ichthyosis
- GeneReviews: http://www.ncbi.nlm.nih.gov/books/NBK1420/
- 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

Patient Information
Patient Name: Donor 8107

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

Test Information
Ordering Physician: [redacted]

Clinic Information: Fairfax Cryobank

Report Date: 05/20/2025

horizon™ natera carrier screen

THYROID DYSHORMONOGENESIS 6

Understanding Your Horizon Carrier Screen Results

What does being a carrier mean?

Your results show that you are a carrier of thyroid dyshormonogenesis 6 (TDH6). Most people with a variant in this gene are carriers of TDH6, but do not have the condition. Some people with a variant in this gene have symptoms of TDH6 as babies that go away as they age.

Your children are at risk for TDH6 or for short-term symptoms of this condition, but you are not certain to have a child with this condition. Further testing can be done to see if your partner or donor is a carrier.

What is thyroid dyshormonogenesis 6 (TDH6)?

TDH6 causes the body to not make enough thyroid hormones, resulting in congenital hypothyroidism (CH).[1,2] Some people with CH have no symptoms. Other people with CH can be less active, sleep more than normal, and have feeding problems or constipation. People with CH that is not treated can also have slow growth and intellectual disability.[2] With early treatment, people with TDH6 usually have normal development.[3] Newborn screening can detect over 90% of babies with CH.[4]

Carriers of TDH6 can have mild hypothyroidism as babies. Thyroid hormone levels can be lower than average at birth and increase with age.[1,2]

Clinical trials involving potential new treatments for this condition could be available (see clinicaltrials.gov).

What causes thyroid dyshormonogenesis 6 (TDH6)?

TDH6 is caused by changes, or variants, in the DUOX2 gene. These changes make the gene not work properly. Genes are a set of instructions inside the cells of our bodies that tell our bodies how to grow and function. Everyone has two copies of the DUOX2 gene. Carriers of TDH6 have one working copy and one non-working copy of the gene. Some carriers have low levels of thyroid hormones as babies, but have normal thyroid function as they get older. People with TDH6 have no working copies of the gene.

TDH6 is usually passed down, or inherited, from both genetic parents. We inherit one copy of the DUOX2 gene from each of our genetic parents. When both genetic parents are carriers, each child has a 1 in 4 (25%) chance of inheriting two non-working genes and having TDH6. Each child also has a 1 in 2 (50%) chance of being a carrier of TDH6 and a 1 in 4 (25%) chance of inheriting two working copies of the gene. This type of inheritance is called autosomal recessive inheritance.

Will my children have thyroid dyshormonogenesis 6 (TDH6)?

If your partner or donor also has a non-working copy of the DUOX2 gene, your children could have TDH6. Each child you have together would have a 1 in 4 (25%) chance of having TDH6. Each child you have together would also have a 1 in 4 (25%) chance of not having any variants in the DUOX2 gene. Each child would have a 1 in 2 (50%) chance of being a carrier and could have symptoms of the condition as a baby.

If your partner or donor has DUOX2 carrier screening and no variants are found, the chance that your children would have two TDH6 variants is very low. In this situation, each child you have together would have a 1 in 2 (50%) chance of being a carrier and could have symptoms of TDH6 as a baby.

What can I do next?

If you want to know if your children are at risk for TDH6, your partner or donor would need to have DUOX2 carrier screening. If you have questions about this testing, please ask your healthcare provider or use the resources below. Many people find it helpful to speak with a genetic counselor.

If your partner or donor is found to be a TDH6 carrier, your children would be at risk for having TDH6. Your children are also at risk of being carriers who have low levels of thyroid hormones as babies.

If you or your partner or surrogate are currently pregnant, tests called CVS (chorionic villus sampling) and amniocentesis can be done during pregnancy to find out if a baby has TDH6. These tests both have a small risk of miscarriage. Babies can also be tested for TDH6 after birth instead.

If you or your partner or surrogate are not yet pregnant, you could have these options:

- natural pregnancy with CVS or amniocentesis to test for TDH6 during pregnancy;
- natural pregnancy and testing the baby after birth for TDH6;
- preimplantation genetic testing (PGT-M) with in vitro fertilization (IVF) to test embryos for TDH6;
- adoption; or
- use of a sperm or egg donor who had no variants found in DUOX2 carrier screening.

Where can I find more information?

- Pediatric Endocrine Society pedsendo.org/patient-resource/congenital-hypothyroidism
- American Thyroid Association thyroid.org/professionals
- CVS marchofdimes.org/chorionic-villus-sampling
- Amniocentesis marchofdimes.org/pregnancy/amniocentesis
- PGT-M natera.com/womens-health/spectrum-preimplantation-genetics

What does this mean for my family?

You likely got (inherited) this non-working gene from one of your genetic parents. Your genetic siblings and other family members could also carry it. You should tell your family members about your test results so they can decide if they want carrier screening for TDH6.

References

1. Moreno JC et al. Inactivating mutations in the gene for thyroid oxidase 2 (THOX2) and congenital hypothyroidism. New Eng. J. Med. 347: 95-102, 2002.
2. Vigone MC et al. Persistent mild hypothyroidism associated with novel sequence variants of the DUOX2 gene in two siblings. Hum. Mutat. 26: 395, 2005.
3. MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US). Congenital hypothyroidism; [updated 2015 Sep 1; cited 2024 March 3]. Available from: https://medlineplus.gov/genetics/condition/congenital-hypothyroidism/.
4. Büyükgebiz A. Newborn screening for congenital hypothyroidism. J Clin Res Pediatr Endocrinol. 2013;5 Suppl 1(Suppl 1):8-12. doi: 10.4274/jcrpe.845. Epub 2012 Nov 15. PMID: 23154158; PMCID: PMC3608007.

VARIANT DETAILS

DUOX2, c.457C>T (p.Q153*), likely pathogenic

- The c.457C>T (p.Q153*) variant in the DUOX2 gene has not been observed in the gnomAD v2.1.1 dataset.
- 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 not been described in ClinVar.

TGM1, c.376C>T (p.R126C), likely pathogenic

- The c.376C>T (p.R126C) variant in the TGM1 gene has been observed at a frequency of 0.0028% 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 autosomal recessive congenital ichthyosis (PMID: 16968736, 19241467).
- This variant has been reported in ClinVar [ID: 39528].

Patient Information
Patient Name: Donor 8107

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

Test Information
Ordering Physician: [redacted]

Clinic Information: Fairfax Cryobank

Report Date: 05/20/2025

horizon™ natera carrier screen

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 (*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-GOUTIÈRES 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-GOUTIÈRES SYNDROME, TREX1-RELATED (*TREX1*) negative
ALPHA-MANNOSIDOSIS (*MAN2B1*) negative
ALPHA-THALASSEMIA (*HBA1/HBA2*) negative
ALPORT SYNDROME, COL4A3-RELATED (*COL4A3*) negative
ALPORT SYNDROME, COL4A4-RELATED (*COL4A4*) negative
ALSTROM SYNDROME (*ALMS1*) negative
AMISH INFANTILE EPILEPSY SYNDROME (*ST3GAL5*) negative
ANDERMANN SYNDROME (*SLC12A6*) negative
ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (*GATM*) negative
ARGININEMIA (*ARG1*) negative
ARGININOSUCCINATE LYASE DEFICIENCY (*ASL*) negative
AROMATASE DEFICIENCY (*CYP19A1*) negative
ASPARAGINE SYNTHETASE DEFICIENCY (*ASNS*) negative
ASPARTYLGLYCOSAMINURIA (*AGA*) negative
ATAXIA WITH VITAMIN E DEFICIENCY (*TTPA*) negative
ATAXIA-TELANGIECTASIA (*ATM*) negative
ATAXIA-TELANGIECTASIA-LIKE DISORDER 1 (*MRE11*) negative
ATRANSFERRINEMIA (*TF*) negative
AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (*SLC35A3*) negative
AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (*AIRE*) negative
AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (ARCI), SLC27A4-RELATED (*SLC27A4*) negative
AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (*SACS*) negative

B
BARDET-BIEDL SYNDROME, ARL6-RELATED (*ARL6*) negative
BARDET-BIEDL SYNDROME, BBS10-RELATED (*BBS10*) negative
BARDET-BIEDL SYNDROME, BBS12-RELATED (*BBS12*) negative
BARDET-BIEDL SYNDROME, BBS1-RELATED (*BBS1*) negative
BARDET-BIEDL SYNDROME, BBS2-RELATED (*BBS2*) negative
BARDET-BIEDL SYNDROME, BBS4-RELATED (*BBS4*) negative
BARDET-BIEDL SYNDROME, BBS5-RELATED (*BBS5*) negative
BARDET-BIEDL SYNDROME, BBS7-RELATED (*BBS7*) negative
BARDET-BIEDL SYNDROME, BBS9-RELATED (*BBS9*) negative
BARDET-BIEDL SYNDROME, TTC8-RELATED (*TTC8*) negative
BARE LYMPHOCYTE SYNDROME, 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 FRONTOPARIETAL 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 (*TSFM*) negative
COMBINED PITUITARY HORMONE DEFICIENCY 1 (*POU1F1*) negative
COMBINED PITUITARY HORMONE DEFICIENCY-2 (*PROP1*) negative
CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (*CYP11B1*) negative
CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (*CYP17A1*) negative
CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (*CYP21A2*) negative
CONGENITAL ADRENAL INSUFFICIENCY, CYP11A1-RELATED (*CYP11A1*) negative
CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (*MPL*) negative
CONGENITAL CHRONIC DIARRHEA (*DGAT1*) negative
CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, ALG1-RELATED (*ALG1*) negative
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-Related (*PMM2*) negative
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (*MPI*) negative
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (*ALG6*) negative
CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (*SEC23B*) negative
CONGENITAL FINNISH NEPHROSIS (*NPHS1*) negative
CONGENITAL HYDROCEPHALUS 1 (*CCDC88C*) negative
CONGENITAL HYPERINSULINISM, KCNJ11-Related (*KCNJ11*) negative
CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (CIPA) (*NTRK1*) negative
CONGENITAL MYASTHENIC SYNDROME, CHAT-RELATED (*CHAT*) negative
CONGENITAL MYASTHENIC SYNDROME, 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 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

Patient Information
Patient Name: Donor 8107

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

Test Information
Ordering Physician: [redacted]

Clinic Information: Fairfax Cryobank

Report Date: 05/20/2025

horizon™ natera carrier screen

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

E
EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY, CAD-RELATED (*CAD*) negative
EHLERS-DANLOS SYNDROME TYPE VI (*PLOD1*) negative
EHLERS-DANLOS SYNDROME, CLASSIC-LIKE, TNXB-RELATED (*TNXB*) negative
EHLERS-DANLOS SYNDROME, TYPE VII C (*ADAMTS2*) negative
ELLIS-VAN CREVELD SYNDROME, EVC2-RELATED (*EVC2*) negative
ELLIS-VAN CREVELD SYNDROME, EVC-RELATED (*EVC*) negative
ENHANCED S-CONE SYNDROME (*NR2E3*) negative
EPIMERASE DEFICIENCY (GALACTOSEMIA TYPE III) (*GALE*) negative
EPIPHYSEAL DYSPLASIA, MULTIPLE, 7/DESBUQUOIS DYSPLASIA 1 (*CANT1*) negative
ERCC6-RELATED DISORDERS (*ERCC6*) negative
ERCC8-RELATED DISORDERS (*ERCC8*) negative
ETHYLMALONIC ENCEPHALOPATHY (*ETHE1*) negative

F
FACTOR XI DEFICIENCY (*F11*) negative
FAMILIAL DYSAUTONOMIA (*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 (*ASAH1*) negative
FOVEAL HYPOPLASIA (*SLC38A8*) negative
FRASER SYNDROME 3, GRIP1-RELATED (*GRIP1*) negative
FRASER SYNDROME, FRAS1-RELATED (*FRAS1*) negative
FRASER SYNDROME, FREM2-RELATED (*FREM2*) negative
FRIEDREICH ATAXIA (*FXN*) negative
FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY (*FBP1*) negative
FUCOSIDOSIS, FUCA1-RELATED (*FUCA1*) negative
FUMARASE DEFICIENCY (*FH*) negative

G
GABA-TRANSAMINASE DEFICIENCY (*ABAT*) negative
GALACTOKINASE DEFICIENCY (GALACTOSEMIA, TYPE II) (*GALK1*) negative
GALACTOSEMIA (*GALT*) negative
GALACTOSIALIDOSIS (*CTSA*) negative
GAUCHER DISEASE (*GBA*) negative
GCH1-RELATED CONDITIONS (*GCH1*) negative
GDF5-RELATED CONDITIONS (*GDF5*) negative
GERODERMA OSTEODYSPLASTICA (*GORAB*) negative
GITELMAN SYNDROME (*SLC12A3*) negative
GLANZMANN THROMBASTHENIA (*ITGB3*) negative
GLUTARIC ACIDEMIA, TYPE 1 (*GCDH*) negative
GLUTARIC ACIDEMIA, TYPE 2A (*ETFA*) negative
GLUTARIC ACIDEMIA, TYPE 2B (*ETFB*) negative
GLUTARIC ACIDEMIA, TYPE 2C (*ETFDH*) negative
GLUTATHIONE SYNTHETASE DEFICIENCY (*GSS*) negative
GLYCINE ENCEPHALOPATHY, AMT-RELATED (*AMT*) negative
GLYCINE ENCEPHALOPATHY, GLDC-RELATED (*GLDC*) negative
GLYCOGEN STORAGE DISEASE TYPE 5 (McArdle Disease) (*PYGM*) negative
GLYCOGEN STORAGE DISEASE TYPE IXB (*PHKB*) negative
GLYCOGEN STORAGE DISEASE TYPE IXC (*PHKG2*) negative
GLYCOGEN STORAGE DISEASE, TYPE 1a (*G6PC*) negative
GLYCOGEN STORAGE DISEASE, TYPE 1b (*SLC37A4*) negative
GLYCOGEN STORAGE DISEASE, TYPE 2 (POMPE DISEASE) (*GAA*) negative
GLYCOGEN STORAGE DISEASE, TYPE 3 (*AGL*) negative
GLYCOGEN STORAGE DISEASE, TYPE 4 (*GBE1*) negative
GLYCOGEN STORAGE DISEASE, TYPE 7 (*PFKM*) negative
GRACILE SYNDROME (*BCS1L*) negative
GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY (*GAMT*) negative

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

K
KRABBE DISEASE (*GALC*) negative

L
LAMELLAR ICHTHYOSIS, TYPE 1 (*TGM1*) see first page

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

M
MALONYL-COA DECARBOXYLASE DEFICIENCY (*MLYCD*) negative
MAPLE SYRUP URINE DISEASE, TYPE 1A (*BCKDHA*) negative
MAPLE SYRUP URINE DISEASE, TYPE 1B (*BCKDHB*) negative
MAPLE SYRUP URINE DISEASE, TYPE 2 (*DBT*) negative
MCKUSICK-KAUFMAN SYNDROME (*MKKS*) negative
MECKEL SYNDROME 7/NEPHRONOPHTHISIS 3 (*NPHP3*) negative
MECKEL-GRUBER SYNDROME, TYPE 1 (*MKS1*) negative
MECR-RELATED NEUROLOGIC DISORDER (*MECR*) negative
MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (*ACADM*) negative
MEDNIK SYNDROME (*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 CBLC (*MMACHC*) negative
METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CblD (*MMADHC*) negative
METHYLMALONIC ACIDURIA, MMAA-RELATED (*MMAA*) negative
METHYLMALONIC ACIDURIA, MMAB-RELATED (*MMAB*) negative
METHYLMALONIC ACIDURIA, TYPE MUT(0) (*MUT*) negative
MEVALONIC KINASE DEFICIENCY (*MVK*) negative
MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II (*PCNT*) negative
MICROPHTHALMIA / ANOPHTHALMIA, VSX2-RELATED (*VSX2*) negative
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, ACAD9-RELATED (*ACAD9*) negative
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFAF5-RELATED (*NDUFAF5*) negative
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFS6-RELATED (*NDUFS6*) negative
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 1 (*NDUFS4*) negative
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 10 (*NDUFAF2*) negative
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 17 (*NDUFAF6*) negative
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 19 (*FOXRED1*) negative
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 3 (*NDUFS7*) negative
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 4 (*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 (MLASA1) (*PUS1*) negative
MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (*HADHB*) negative
MOLYBDENUM COFACTOR DEFICIENCY TYPE B (*MOCS2*) negative
MOLYBDENUM COFACTOR DEFICIENCY, TYPE A (*MOCS1*) 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 (*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, TMPRSS3-RELATED (*TMPRSS3*) negative
NONSYNDROMIC INTELLECTUAL DISABILITY (*CC2D1A*) negative
NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (*SAMD9*) negative

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

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

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) (*ATP8B1*) negative
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 2 (*ABCB11*) negative
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 4 (PFIC4) (*TJP2*) negative
PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA (*CCN6*) negative
PROLIDASE DEFICIENCY (*PEPD*) negative
PROPIONIC ACIDEMIA, PCCA-RELATED (*PCCA*) negative
PROPIONIC ACIDEMIA, PCCB-RELATED (*PCCB*) negative
PSEUDOXANTHOMA ELASTICUM (*ABCC6*) negative
PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (PCD) DEFICIENCY (*PCBD1*) negative
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
RLBP1-RELATED RETINOPATHY (*RLBP1*) negative
ROBERTS SYNDROME (*ESCO2*) negative
RYR1-RELATED CONDITIONS (*RYR1*) negative

S
SALLA DISEASE (*SLC17A5*) negative
SANDHOFF DISEASE (*HEXB*) negative
SCHIMKE IMMUNOOSSEOUS DYSPLASIA (*SMARCAL1*) negative
SCHINDLER DISEASE (*NAGA*) negative
SEGAWA SYNDROME, TH-RELATED (*TH*) negative
SENIOR-LOKEN SYNDROME 4/NEPHRONOPHTHISIS 4 (*NPHP4*) negative
SEPIAPTERIN REDUCTASE DEFICIENCY (*SPR*) negative
SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3D-RELATED (*CD3D*) negative
SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3E-RELATED (*CD3E*) negative
SEVERE COMBINED IMMUNODEFICIENCY (SCID), FOXN1-RELATED (*FOXN1*) negative
SEVERE COMBINED IMMUNODEFICIENCY (SCID), IKBKB-RELATED (*IKBKB*) negative
SEVERE COMBINED IMMUNODEFICIENCY (SCID), IL7R-RELATED (*IL7R*) negative
SEVERE COMBINED IMMUNODEFICIENCY (SCID), JAK3-RELATED (*JAK3*) negative
SEVERE COMBINED IMMUNODEFICIENCY (SCID), PTPRC-RELATED (*PTPRC*) negative
SEVERE COMBINED IMMUNODEFICIENCY (SCID), RAG1-RELATED (*RAG1*) negative
SEVERE COMBINED IMMUNODEFICIENCY, ADA-Related (*ADA*) negative
SEVERE COMBINED IMMUNODEFICIENCY, TYPE ATHABASKAN (*DCLRE1C*) negative
SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY (*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
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*) see first page
TRANSCOBALAMIN II DEFICIENCY (*TCN2*) negative
TRICHOHEPATOENTERIC SYNDROME, SKIC2-RELATED (*SKIC2*) negative
TRICHOHEPATOENTERIC SYNDROME, TTC37-RELATED (*TTC37*) negative
TRICHOTHIODYSTROPHY 1/XERODERMA PIGMENTOSUM, GROUP D (*ERCC2*) negative
TRIMETHYLAMINURIA (*FMO3*) negative
TRIPLE A SYNDROME (*AAAS*) negative
TSHR-RELATED CONDITIONS (*TSHR*) negative
TYROSINEMIA TYPE III (*HPD*) negative
TYROSINEMIA, TYPE 1 (*FAH*) negative
TYROSINEMIA, TYPE 2 (*TAT*) negative

U
USHER SYNDROME, TYPE 1B (*MYO7A*) negative
USHER SYNDROME, TYPE 1C (*USH1C*) negative
USHER SYNDROME, TYPE 1D (*CDH23*) negative
USHER SYNDROME, TYPE 1F (*PCDH15*) negative
USHER SYNDROME, TYPE 1J/DEAFNESS, AUTOSOMAL RECESSIVE, 48 (*CIB2*) negative
USHER SYNDROME, TYPE 2A (*USH2A*) negative
USHER SYNDROME, TYPE 2C (*ADGRV1*) negative
USHER SYNDROME, TYPE 3 (*CLRN1*) negative

V
VERY LONG-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (*ACADVL*) negative
VICI SYNDROME (*EPG5*) negative
VITAMIN D-DEPENDENT RICKETS, TYPE 1A (*CYP27B1*) negative
VITAMIN D-RESISTANT RICKETS TYPE 2A (*VDR*) negative
VLDLR-ASSOCIATED CEREBELLAR HYPOPLASIA (*VLDLR*) negative

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

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

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

Patient Information
Patient Name: Donor 8107

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

Test Information
Ordering Physician: [redacted]

Clinic Information: Fairfax Cryobank

Report Date: 05/20/2025

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

Testing Methodology, Limitations, and Comments:

Next-generation sequencing (NGS)

Sequencing library prepared from genomic DNA isolated from a patient sample is enriched for targets of interest using standard hybridization capture protocols and PCR amplification (for targets specified below). NGS is then performed to achieve the standards of quality control metrics, including a minimum coverage of 99% of targeted regions at 20X sequencing depth. Sequencing data is aligned to human reference sequence, followed by deduplication, metric collection and variant calling (coding region +/- 20bp). Variants are then classified according to ACMGG/AMP standards of interpretation using publicly available databases including but not limited to ENSEMBL, HGMD Pro, ClinGen, ClinVar, 1000G, ESP and gnomAD. Variants predicted to be pathogenic or likely pathogenic for the specified diseases are reported. It should be noted that the data interpretation is based on our current understanding of the genes and variants at the time of reporting. Putative positive sequencing variants that do not meet internal quality standards or are within highly homologous regions are confirmed by Sanger sequencing or gene-specific long-range PCR as needed prior to reporting.

Copy Number Variant (CNV) analysis is limited to deletions involving two or more exons for all genes on the panel, in addition to specific known recurrent single-exon deletions. CNVs of small size may have reduced detection rate. This method does not detect gene inversions, single-exonic and sub-exonic deletions (unless otherwise specified), and duplications of all sizes (unless otherwise specified). Additionally, this method does not define the exact breakpoints of detected CNV events. Confirmation testing for copy number variation is performed by specific PCR, Multiplex Ligation-dependent Probe Amplification (MLPA), next generation sequencing, or other methodology.

This test may not detect certain variants due to local sequence characteristics, high/low genomic complexity, homologous sequence, or allele dropout (PCR-based assays). Variants within noncoding regions (promoter, 5'UTR, 3'UTR, deep intronic regions, unless otherwise specified), small deletions or insertions larger than 25bp, low-level mosaic variants, structural variants such as inversions, and/or balanced translocations may not be detected with this technology.

SPECIAL NOTES

For ABCC6, sequencing variants in exons 1-7 are not detected due to the presence of regions of high homology.

For CFTR, when the CFTR R117H variant is detected, reflex analysis of the polythymidine variations (5T, 7T and 9T) at the intron 9 branch/acceptor site of the CFTR gene will be performed. Multi-exon duplication analysis is included.

For CYP21A2, targets were enriched using long-range PCR amplification, followed by next generation sequencing. Duplication analysis will only be performed and reported when c.955C>T (p.Q319*) is detected. Sequencing and CNV analysis may have reduced sensitivity, if variants result from complex rearrangements, in trans with a gene deletion, or CYP21A2 gene duplication on one chromosome and deletion on the other chromosome. This analysis cannot detect sequencing variants located on the CYP21A2 duplicated copy.

For DDX11, sequencing variants in exons 7-11 and CNV for the entire gene are not analyzed due to high sequence homology.

For GJB2, CNV analysis of upstream deletions of GJB6-D13S1830 (309kb deletion) and GJB6-D13S1854 (232kb deletion) is included.

For HBA1/HBA2, CNV analysis is offered to detect common deletions of -alpha3.7, -alpha4.2, --MED, --SEA, --FIL, --THAI, --alpha20.5, and/or HS-40.

For OTOA, sequencing variants in exons 25-29 and CNV in exons 21-29 are not analyzed due to high sequence homology.

For RPGRIP1L, variants in exon 23 are not detected due to assay limitation.

For SAMD9, only p.K1495E variant will be analyzed and reported.

Friedreich Ataxia (FXN)

The GAA repeat region of the FXN gene is assessed by trinucleotide PCR assay and capillary electrophoresis. Variances of +/-1 repeat for normal alleles and up to +/-3 repeats for premutation alleles may occur. For fully penetrant expanded alleles, the precise repeat size cannot be determined, therefore the approximate allele size is reported. Sequencing and copy number variants are analyzed by next-generation sequencing analysis.

Friedreich Ataxia Repeat Categories

Categories	GAA Repeat Sizes
Normal	<34
Premutation	34 - 65
Full	>65

Patient Information
Patient Name: Donor 8107

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

Test Information
Ordering Physician: [redacted]

Clinic Information: Fairfax Cryobank

Report Date: 05/20/2025

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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.