

# **Donor 7156**

# **Genetic Testing Summary**

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

Last Updated: 12/16/24

Donor Reported Ancestry: Cuban 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.	Carrier: Thyroid Dyshormonogenesis 6 (DUOX2)  Carrier: Walker-Warburg Syndrome, CRPPA-Related  Carrier: Friedreich Ataxia One expanded size of approximately 210 (GAA)  Negative for other genes sequenced.	Partner testing is recommended before using this donor.

<sup>\*</sup>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.

<sup>\*\*</sup>Donor residual risk is the chance the donor is still a carrier after testing negative.

Patient Name: Donor 7156

Date Of Birth:

Gender: Male

Ethnicity: Hispanic/Latin American

N/A

Patient ID:

Medical Record #: N/A

Collection Kit: Accession ID:

Case File ID:

**Test Information** 

Ordering Physician:

Clinic Information: Fairfax Cryobank

Phone:

Report Date: Sample Collected: Sample Received:

Sample Type:

horizon<sup>™</sup>

## 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:**



10/18/2024

10/03/2024

10/04/2024

Blood

# **CARRIER for Thyroid Dyshormonogenesis 6**

Positive for the likely pathogenic variant c.4552G>A (p.G1518S) 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.

# **CARRIER for Walker-Warburg Syndrome, CRPPA-Related**

Positive for the likely pathogenic variant c.1250del (p.Q417Rfs\*10) in the CRPPA gene. If this individual's partner is a carrier for WALKER-WARBURG SYNDROME, CRPPA-RELATED, 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 Friedreich Ataxia**

One expanded size of approximately 210 GAA repeat allele and one normal allele were detected in the FXN gene. If this individual's partner is a carrier for FRIEDREICH ATAXIA, 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 546 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 <a href="https://www.natera.com/panel-option/h-all/">https://www.natera.com/panel-option/h-all/</a>. 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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Yang Wang, Ph.D., FACMG

J. Dianne Keen-Kim, Ph.D., FACMGG



Patient Name: Donor 7156

Test Information

Ordering Physician:

Clinic Information: Fairfax Cryobank



Date Of Birth: Case File ID:



Report Date:

10/18/2024

#### FRIEDREICH ATAXIA

#### **Understanding Your Horizon Carrier Screen Results**

#### What is Friedreich ataxia?

Friedreich ataxia is an inherited condition that causes problems with movement (ataxia) that worsen over time. Symptoms often start in early to late childhood. People with Friedreich ataxia develop coordination and balance problems, muscle weakness and stiffness (spasticity), and loss of sensation in the arms and legs. The ability to walk is affected and over time some people will need the use of a wheelchair. Other symptoms may include difficulties with speech and gradual loss of vision and/or hearing. An enlarged, weakened heart muscle (hypertrophic cardiomyopathy) is found in about two-thirds of people with Friedreich ataxia, and diabetes occurs in about a third.

Some people have a late-onset form of Friedreich ataxia with symptoms that do not start until after age 25. Rarely, a very late-onset form occurs, with symptoms starting after age 40. These later-onset forms have symptoms that progress more slowly than the typical childhood-onset form. Currently there is no cure for Friedreich ataxia, and treatment is based on symptoms. Clinical trials involving potential new treatments for this condition may be available (see clinicaltrials.gov).

#### What causes Friedreich ataxia?

Friedreich ataxia is caused by a gene change, or variant, in both copies of the FXN gene pair. The variant in the FXN gene pair that usually causes Friedreich ataxia is called a GAA expansion, though other rare variants can also cause Friedreich ataxia. Humans typically have 33 or fewer copies of GAA in each FXN gene. People with Friedreich ataxia typically have 66 or more copies of GAA in each of their two FXN genes, which is called an expanded, or full penetrance, variant. The large number of repeated GAAs cause the gene to not work properly or not work at all. A small number of people with Friedreich ataxia have a GAA expansion in one copy of their FXN gene and a different kind of variant in their other copy of the FXN gene, which stops it from working correctly. When both copies of the FXN gene do not work correctly, it leads to the symptoms described above. Friedreich ataxia is inherited in an autosomal recessive manner. This means that, in most cases, both parents of a child with Friedreich ataxia are carriers of an expanded GAA repeat are usually healthy and do not have Friedreich ataxia themselves. If the mother and father are both carriers of an expanded GAA repeat, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their FXN GAA repeat variants to the child, who will then have the condition.

There have been some reports of people with an intermediate GAA repeat size of 34 to 65 in one of their FXN genes. A GAA repeat size of 34 to 65 is called a premutation. An FXN premutation can expand to a larger GAA repeat size when passed to a child. If one parent has an expanded FXN variant and the other parent has a premutation, there is a less than 25% chance in each pregnancy to have a child with Friedreich ataxia.

People found to carry more than one variant for Friedreich ataxia should discuss their risk for having an affected child and any potential risks to their own health with their healthcare 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 (<a href="https://www.nsgc.org">www.nsgc.org</a>).

Your siblings and other relatives are at increased risk to also have this variant. 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 Friedreich ataxia ordered by a healthcare professional. If your partner is not found to be a carrier of Friedreich ataxia, your risk of having an affected child is greatly reduced. If your partner is found to be a carrier, you can consider having prenatal diagnostic testing done through chorionic villus sampling (CVS) or amniocentesis during pregnancy or can choose to have the baby tested after birth for Friedreich ataxia.

If you are not yet pregnant, your partner can have carrier screening for Friedreich ataxia ordered by a healthcare professional. If your partner is found to be a carrier of Friedreich ataxia, you have several reproductive options to consider:

- natural pregnancy with or without prenatal diagnostic testing of the fetus or testing the baby after birth for Friedreich ataxia;
- preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for Friedreich ataxia; or
- adoption or use of a sperm or egg donor who is not a carrier of Friedreich ataxia.

# What resources are available?

- MedlinePlus Genetics medlineplus.gov/genetics/condition/friedreich-ataxia
- GeneReviews www.ncbi.nlm.nih.gov/books/NBK1281
- CVS <u>www.marchofdimes.org/find-support/topics/planning-baby/chorionic-villus-sampling</u>
- Amniocentesis www.marchofdimes.org/find-support/topics/planning-baby/amniocentesis
- PGD with IVF <u>www.natera.com/spectrum</u>



Patient	Information
Patient I	Name:

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#### 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 <u>pedsendo.org/patient-resource/congenital-hypothyroidism</u>
- American Thyroid Association <a href="mailto:thyroid.org/professionals">thyroid.org/professionals</a>
- CVS marchofdimes.org/chorionic-villus-sampling
- Amniocentesis <u>marchofdimes.org/pregnancy/amniocentesis</u>
- PGT-M natera.com/womens-health/spectrum-preimplantation-genetics

# What does this mean for my family?



Patient Info	ormation
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Patient	Name:	

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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: <a href="https://medlineplus.gov/genetics/condition/congenital-hypothyroidism/">https://medlineplus.gov/genetics/condition/congenital-hypothyroidism/</a>.

  4. Büyükgebiz A. Newborn screening for congenital hypothyroidism. J Clin Res Pediatr Endocrinol. 2013;5 Suppl 1(Suppl 1):8-12. doi: <a href="https://medlineplus.gov/genetics/condition/congenital-hypothyroidism/">10.4274/jcrpe.845</a>. Epub 2012 Nov 15. PMID: 23154158; PMCID: PMC3608007.



<b>Patient Information</b>
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#### WALKER-WARBURG SYNDROME, CRPPA-RELATED

#### **Understanding Your Horizon Carrier Screen Results**

## What is Walker-Warburg Syndrome, CRPPA-Related?

Walker-Warburg Syndrome, CRPPA-Related is an inherited disorder that affects many parts of the body, especially the brain, eyes, and muscles. Signs and symptoms are often present before birth but sometimes start in infancy and include weak muscle tone (hypotonia), excess fluid on the brain (hydrocephalus), severe brain abnormalities, and eye defects with vision problems. Infants and children with Walker-Warburg Syndrome, CRPPA-Related have worsening muscle weakness, problems with movement and coordination, seizures, and severe developmental delay with intellectual disability. Although symptoms vary from person to person, lifespan is usually shortened with death often occurring in early childhood. Currently, there is no cure or specific treatment for this disorder. Clinical trials involving potential new treatments for this condition may be available (see <a href="https://www.clinicaltrials.gov">www.clinicaltrials.gov</a>). Rarely, mutations in the same gene pair cause a related condition called Limb-Girdle Muscular Dystrophy, Type 2U. Limb-Girdle Muscular Dystrophy, Type 2U causes severe muscle weakness in the shoulder and hip areas along with muscle pain during exertion that usually starts in childhood. The information below is about Walker-Warburg Syndrome, CRPPA-Related, the more common condition. However, the inheritance pattern and reproductive options listed below apply to Limb-Girdle Muscular Dystrophy, Type 2U as well.

# What causes Walker-Warburg Syndrome, CRPPA-Related?

Walker-Warburg Syndrome, CRPPA-Related is caused by a change, or mutation, in both copies of the CRPPA (ISPD) gene pair. These mutations cause the genes to not work properly or not work at all. When both copies of the CRPPA (ISPD) gene do not work correctly, it leads to the symptoms described above. It is sometimes, but not always, possible to determine whether a specific mutation in the CRPPA (ISPD) gene will cause Walker-Warburg Syndrome, CRPPA-Related or Limb-Girdle Muscular Dystrophy, Type 2U. Walker-Warburg Syndrome, CRPPA-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 CRPPA (ISPD) gene to have a child with Walker-Warburg Syndrome, CRPPA-Related. People who are carriers for Walker-Warburg Syndrome, CRPPA-Related are usually healthy and do not have symptoms, 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 Walker-Warburg Syndrome, CRPPA-Related or related condition there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their CRPPA (ISPD) gene mutations to the child, who will then have this disorder. Individuals found to carry more than one mutation for Walker-Warburg Syndrome, CRPPA-Related should discuss their risk for having an affected child and any potential effects to their own health with their health care provider. There are a number of other forms of Walker-Warburg Syndrome and Limb-Girdle Muscular Dystrophy, each caused by mutations in different genes. A person who carries a mutation in the CRPPA gene is not likely to be at increased risk for having children with the other forms of these disorders.

#### What can I do next?

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website (<a href="www.nsgc.org">www.nsgc.org</a>). 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 Walker-Warburg Syndrome, CRPPA-Related ordered by a health care professional. If your partner is not found to be a carrier for Walker-Warburg Syndrome, CRPPA-Related, your risk of having an affected child is greatly reduced. Couples at risk of having a baby with Walker-Warburg Syndrome, CRPPA-Related 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 Walker-Warburg Syndrome, CRPPA-Related ordered by a health care professional. If your partner is found to be a carrier for Walker-Warburg Syndrome, you have several reproductive options to consider:

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

## What resources are available?

- Genetics Home Reference: http://ghr.nlm.nih.gov/condition/walker-warburg-syndrome
- Prenatal diagnosis done through CVS: <a href="http://www.marchofdimes.org/chorionic-villus-sampling.aspx">http://www.marchofdimes.org/chorionic-villus-sampling.aspx</a>
- Prenatal diagnosis done through Amniocentesis: <a href="http://www.marchofdimes.org/amniocentesis.aspx">http://www.marchofdimes.org/amniocentesis.aspx</a>
- PGD with IVF: http://www.natera.com/spectrum



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# **VARIANT DETAILS**

# CRPPA, c.1250del (p.Q417Rfs\*10), likely pathogenic

- The c.1250del (p.Q417Rfs\*10) variant in the CRPPA gene has not been observed in the gnomAD v2.1.1 dataset.
- This premature termination variant is predicted to escape nonsense-mediated decay (NMD) but impact a significant portion of the protein length or a critical region of the protein, potentially disrupting normal protein function.
- This variant has not been described in ClinVar.

# DUOX2, c.4552G>A (p.G1518S), likely pathogenic

- The c.4552G>A (p.G1518S) variant in the DUOX2 gene has been observed at a frequency of 0.0075% 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 thyroid dyshormonogenesis 6 (PMID: 20187165, 31030636).
- Functional studies demonstrated that this variant causes reduced H2O2-producing activities (PMID: 20187165).
- This variant has been reported in ClinVar [ID: 1210930].



Patient Name:

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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**

17-BETA HYDROXYSTEROID DEHYDROGENASE 3 DEFICIENCY (HSD17B3) negative

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-ALPHA-REDUCTASE DEFICIENCY (SRD5A2) negative

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

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, RNASEH/2B-RELATED (RNASEH/2B) negative AICARDI-GOUTIERES SYNDROME, RNASEH/2C-RELATED (RNASEH/2C) 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 (573GAL5) negative
ANDERMANN SYNDROME (SLC12A6) negative

ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY)

(GATM) negative
ARGININEMIA (ARG1) negative
ARGININOSUCCINATE LYASE DEFICIENCY (ASL) negative

ARGINIOSOCCINATE L'IASE 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

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 BART 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

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 (VP513A) 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

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 HYPERINSULINISM, RCNJ11-Related (RCNJ11) 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-BIFUNCTIONAL PROTEIN DEFICIENCY (HSD17B4) negative



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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

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, 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

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 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 F (FANCF) negative FANCONI ANEMIA, GROUP I (FANCG) negative FANCONI ANEMIA, GROUP J (BRIP1) negative FANCONI ANEMIA, GROUP L (FANCL) 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) see first page FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY (FBP1) negative

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 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
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 IXE (PHKB) negative
GLYCOGEN STORAGE DISEASE TYPE IXC (PHKG2) 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

FUCOSIDOSIS, FUCA1-RELATED (FUCA1) negative FUMARASE DEFICIENCY (FH) negative

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

HARLEQUIN ICHTHYOSIS (ABCA12) negative
HEME OXYGENASE 1 DEFICIENCY (HMOX1) negative

HEMOCHROMATOSIS TYPE 2A (HFE2) negative HEMOCHROMATOSIS, TYPE 3, TFR2-Related (TFR2) negative
HEPATOCEREBRAL MITOCHONDRIAL DNA DEPLETION SYNDROME, MPV17-RELATED (MPV17) negative HEREDITARY FRUCTOSE INTOLERANCE (ALDOB) negative HEREDITARY HEMOCHROMATOSIS TYPE 2B (HAMP) negative HEREDITARY SPASTIC PARAPARESIS, TYPE 49 (TECPR2) negative HEREDITARY SPASTIC PARAPARESIS, 17PE 49 (TECPK2) negative HEREDITARY SPASTIC PARAPLEGIA, CYP7B1-RELATED (CYP7B1) negative HERMANSKY-PUDLAK SYNDROME, AP3B1-RELATED (BLOC1S3) 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, 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 AND MEGALOBLASTIC ANEMIA TYPE CBLG (MTR)
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

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

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, ITGAG-RELATED (ITGAG) 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

KRABBE DISEASE (GALC) negative

LAMELLAR ICHTHYOSIS, TYPE 1 (TGM1) negative



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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 II A (SANFILIPPO A) (SGSH) negative MUCOPOLYSACCHARIDOSIS, TYPE III A (SANFILIPPO B) (NAGLU) 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 IV (HYAL1) negative

MUCOPOLYSACCHARIDOSIS, TYPE VI (MAROTEAUX-LAMY) (ARSB) negative MUCOPOLYSACCHARIDOSIS, TYPE VI (MAROTEAUX-LAMY) (ARSB) negative MUCOPOLYSACCHARIDOSIS, TYPE VII (GUSB) negative MULIBREY NANISM (TRIM37) negative MULIBREY PRENGLIM 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-ACETYLGLUTAMATE SYNTHASE DEFICIENCY (NAGS) negative

N-ACETYLGLOTAMATE 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, CLNS-RELATED (CLN8) negative NEURONAL CEROID LIPOFUSCINOSIS, MFSD8-RELATED (MFSD8) negative NEURONAL CEROID LIPOFUSCINOSIS, PPT1-RELATED (PPT1) negative

NEURONAL CEROID LIPOFUSCINOSIS, PP11-RELATED (PP11) 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 NIMEGEN 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

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

PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION (PANK2) negative PAPILLON LEFÈVRE SYNDROME (CTSC) negative PARKINSON DISEASE 15 (FBXO7) negative PENDRED SYNDROME (SLC26A4) negative PENDRED SYNDROME (SLCZOA4) 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

# 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 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 2F (SGCD) negative LIMB-GIRDLE DELIVERAGE AND ASSESSED ASSESSED AND ASSESSED ASSESS

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

MALONYL-COA DECARBOXYLASE DEFICIENCY (MLYCD) negative MAPLE SYRUP URINE DISEASE, TYPE 1A (BCKDHA) negative MAPLE SYRUP URINE DISEASE, TYPE 1B (BCKDHB) negative MAPLE STRUP URINE DISEASE, TYPE 2 (DBT) 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 (MK51) 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 CBLC (MMADHC) negative METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CBID (MMADHC) negative

METHYLMALONIC ACIDURIA, MMAA-RELATED (MMAA) negative METHYLMALONIC ACIDURIA, MMAB-RELATED (MMAB) negative METHYLMALONIC ACIDURIA, TYPE MUT(0) (MUT) negative

MEVALONIC KINASE DEFICIENCY (MVK) negative
MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II (PCNT) negative
MICROPHTHALMIA / ANOPHTHALMIA, VSX2-RELATED (VSX2) negative

MITOCHONDRIAL COMPLEX 1 DEFICIENCY, ACAD9-RELATED (ACAD9) negative MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFAF5-RELATED (NDUFAF5) negative MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFS6-RELATED (NDUFS6) negative MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 1 (NDUFS4) negative

MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 10 (NDUFAF2) negative MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 17 (NDUFAF6) negative MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 19 (FOXRED1) negative

MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 3 (NDUFST) 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

Patient Name:

**Test Information** 

Ordering Physician:



Date Of Birth:

Case File ID:

Clinic Information:

Report Date:

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 1A (WAT) Hegative PONTOCEREBELLAR HYPOPLASIA, TYPE 2D (SEPSECS) negative PONTOCEREBELLAR HYPOPLASIA, VP553-RELATED (VP553) 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 (GRIPR) negative
PRIMARY HYPEROXALURIA, TYPE 3 (HOGA1) negative
PRIMARY MICROCEPHALY 1, AUTOSOMAL RECESSIVE (MCPH1) negative PROGRESSIVE EARLY-ONSET ENCEPAHLOPATHY 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

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
PSEUDOXANTHOMA ELASTICUM (ABCC6) negative
PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (PCD) DEFICIENCY (PCBD1) negative
PYCNODYSOSTOSIS (CTSK) negative
PYRIDOXAL 5"-PHOSPHATE-DEPENDENT EPILEPSY (PNPO) 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

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

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

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

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

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

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

WALKER-WARBURG SYNDROME, CRPPA-RELATED (CRPPA) see first page 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

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, PEXS-RELATED (PEXS) 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 (PEX26) negative



SMITH-LEMLI-OPITZ SYNDROME (DHCR7) negative SPASTIC PARAPLEGIA, TYPE 15 (ZFYVE26) negative

Patient Name:

Test Information
Ordering Physician:



Date Of Birth:

Case File ID:

Report Date:

Clinic Information:

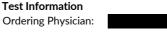
 ${\bf Z}$  ZELLWEGER SPECTRUM DISORDERS, PEX6-RELATED (PEX6)  $\,$  negative

<b>Patient</b>	Information
D	N.I.

Patient Name:

Case File ID:

Date Of Birth:



Clinic Information:





Report Date:

# 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 genespecific 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 singleexon 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, variants in exons 1-9 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.

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, only NM 030653.3:c.1763 - 1G > C variant will be analyzed and reported.

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, variants in exons 20 - 28 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	Test Information	
Patient Name:	Ordering Physician:	
	Clinic Information:	
Date Of Birth:		
Case File ID:		



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.

Report Date:

Ethnicity	Two SMN1 copies carrier risk before g.27134T>G testing	Carrier risk after g.27134T	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 <a href="https://www.natera.com/panel-option/h-all/">https://www.natera.com/panel-option/h-all/</a> 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.





# 7156,DONOR **▲**

DOB Age:
Sex: M Fasting:
Phone:
Patient ID: 7156

Specimen:
Requisition:
Lab Reference ID:
Report Status: FINAL / SEE REPORT

Collected: 10/03/2024 10:20 Received: 10/04/2024 14:29 Reported: 10/13/2024 15:04



# **▲** Hemoglobinopathy Evaluation



Lab: AMD

Value				
,		FINAL		
5.06	Reference Range: 4.20-5.80 Mill/uL	FINAL		
16.2	Reference Range: 13.2-17.1 g/dL	FINAL		
		FINAL		
51.2 H	Reference Range: 38.5-50.0 %	FINAL		
101.2 H	Reference Range: 80.0-100.0 fL	FINAL		
32.0	Reference Range: 27.0-33.0 pg	FINAL		
11.6	Reference Range: 11.0-15.0 %	FINAL		
		FINAL		
97.0	Reference Range: >96.0 %	FINAL		
0.0	Reference Range: <2.0 %	FINAL		
3.0	Reference Range: 2.0-3.2 %	FINAL		
		FINAL		
	5.06 16.2 51.2 H 101.2 H 32.0 11.6 97.0 0.0	5.06 Reference Range: 4.20-5.80 Mill/uL  16.2 Reference Range: 13.2-17.1 g/dL  51.2 H Reference Range: 38.5-50.0 %  101.2 H Reference Range: 80.0-100.0 fL  32.0 Reference Range: 27.0-33.0 pg  11.6 Reference Range: 11.0-15.0 %  97.0 Reference Range: >96.0 %  0.0 Reference Range: <2.0 %		

NORMAL PATTERN

There is a normal pattern of hemoglobins and normal levels of Hb A2 and Hb F are present. No variant hemoglobins are observed. This is consistent with A/A phenotype.

If iron deficiency coexists with a mild/silent beta thalassemia trait Hb A2 may be in the normal range. Rare variant 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.



Chromosome Analysis, Blood

Order ID:

(FINAL)

Specimen Type:

Blood

Clinical Indication:

Gamete donor

**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:

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.

Christine A. Curtis, Ph.D., FACMG, Technical Director, Cytogenetics and Genomics, 703-802-7156, [site AMD10]

Electronic Signature:

10/13/2024 2:20 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

Triority Out of Range A Out of Range PEND Pending Result PRE Preliminary Result FINAL Final Result RE Reissued Result

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