



Donor 7428

Genetic Testing Summary

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

Last Updated: 12/10/24

Donor Reported Ancestry: Austian, Polish, German, Italian

Jewish Ancestry: Yes

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.	<p>Carrier: Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency (CYP21A2)</p> <p>Carrier: Cystic Fibrosis (CFTR)</p> <p>Carrier: Polycystic Kidney Disease, Autosomal Recessive (PKHD1)</p> <p>Carrier: Wilson Disease (ATP7B)</p> <p>Negative for other genes sequenced.</p>	Partner testing is recommended before using this donor.

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

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

Patient Information

Patient Name: Donor 7428

Date Of Birth: [REDACTED]

Gender: Male

Ethnicity: Northern European
Caucasian

Patient ID: N/A

Medical Record #: N/A

Collection Kit: [REDACTED]

Accession ID: N/A

Case File ID: [REDACTED]

Test Information

Ordering Physician: [REDACTED]

Clinic Information: Fairfax Cryobank

Phone: [REDACTED]

Report Date: 07/16/2024

Sample Collected: 07/02/2024

Sample Received: 07/03/2024

Sample Type: Blood

**CARRIER SCREENING REPORT**

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

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

FINAL RESULTS SUMMARY:**CARRIER for Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency**

Positive for the pathogenic variant c.844G>T (p.V282L) [Legacy name: V281L] in the CYP21A2 gene. This variant has been reported in a homozygous state or in conjunction with another variant in individual(s) with non-classic congenital adrenal hyperplasia (PMID: 19263525, 25041270, 32616876). If this individual's partner is a carrier for CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY, their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is recommended.

CARRIER for Cystic Fibrosis

Positive for the pathogenic variant c.1652G>A (p.G551D) in the CFTR gene. A small number of Cystic Fibrosis (CF) carriers may have mild respiratory or other CF-related symptoms. If this individual's partner is a carrier for Cystic Fibrosis, their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

CARRIER for Polycystic Kidney Disease, Autosomal Recessive

Positive for the likely pathogenic variant c.9107T>G (p.V3036G) in the PKHD1 gene. If this individual's partner is a carrier for POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE, 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 Wilson Disease

Positive for the pathogenic variant c.3207C>A (p.H1069Q) in the ATP7B gene. If this individual's partner is a carrier for WILSON DISEASE, their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

Negative for 545 out of 549 diseases

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

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

RECOMMENDATIONS

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

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Linyan Meng, Ph.D.
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Laboratory Director, Natera

Patient Information

Patient Name: Donor 7428

Test Information

Ordering Physician: [REDACTED]



Clinic Information: Fairfax Cryobank

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: 07/16/2024

CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY

Understanding Your Horizon Carrier Screen Results

What is Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency?

Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency (also called 21-Hydroxylase Deficiency) is an inherited disorder that causes the adrenal glands, the organs that sit on top of the kidneys, to make decreased amounts of the hormones cortisol and aldosterone and increased amounts of male sex hormones called androgens.

There are three forms of 21-Hydroxylase Deficiency. The most common and severe form is called the 'salt-wasting type' with signs and symptoms that are often present at birth. Babies with the salt-wasting type of 21-Hydroxylase Deficiency are at risk for losing large amounts of sodium in the urine due to too low a level of aldosterone hormone. These 'salt-wasting crises' can lead to poor feeding, weight loss, dehydration, vomiting, low blood pressure, and shock, and can be life-threatening if not treated quickly. Symptoms in females include being born with external genitals that do not have the typical appearance of male or female (ambiguous genitalia). Over time, affected females may also have early puberty, rapid early growth with short adult height, increased body hair (hirsutism), male pattern baldness, irregular menstrual periods, and decreased fertility. Affected males have normal genitals at birth but are at risk for salt-wasting crises and may have increased penis size and decreased testicle size over time as well as an early growth spurt with short adult height. Some males with this form have decreased fertility due to benign growths in their testicles called 'testicular adrenal rest tumors' (TART).

The 'simple virilizing type' of 21-Hydroxylase Deficiency has similar symptoms to the salt-wasting type except babies with the simple virilizing type are not at risk for salt wasting crises.

The mildest form of 21-Hydroxylase Deficiency is called the 'non-classical type'. People with the nonclassical type of 21-Hydroxylase Deficiency have normal external genitals. Signs and symptoms may begin as early as childhood or not until adulthood and may include an early growth spurt with short adult height, early puberty, and acne. Additional symptoms in females may include excess body hair, male pattern baldness, irregular periods, and decreased fertility. Additional symptoms in males may include early and heavy facial hair and small testicles. Some people with this type never develop symptoms.

Currently, there is no cure for 21-Hydroxylase Deficiency. However, hormone replacement therapy can prevent or lessen some or all of the symptoms. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency?

21-Hydroxylase Deficiency is caused by a change, or mutation, in both copies of the CYP21A2 gene pair. These mutations cause the genes to not work properly or not work at all. The function of the CYP21A2 genes is to help make sex hormones and other hormones. When both copies of this gene do not work correctly, it leads to the symptoms described above.

21-Hydroxylase Deficiency 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 CYP21A2 gene to have a child with 21-Hydroxylase Deficiency. People who are carriers for 21-Hydroxylase Deficiency 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 21-Hydroxylase Deficiency, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their CYP21A2 gene mutations to the child, who will then have this condition. It is sometimes, but not always, possible to determine whether a specific mutation in the CYP21A2 gene will cause the salt-wasting type, the simple virilizing type, or the non-classic type of 21-Hydroxylase Deficiency.

Individuals found to carry more than one mutation for 21-Hydroxylase Deficiency 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 Congenital Adrenal Hyperplasia, each caused by mutations in different genes. A person who is a carrier for Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency is not likely to be at increased risk for having a child 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 21-Hydroxylase Deficiency ordered by a health care professional. If your partner is not found to be a carrier for 21-Hydroxylase Deficiency, your risk of having an affected child is greatly reduced. Couples at risk of having a baby with 21-Hydroxylase Deficiency 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 21-Hydroxylase Deficiency ordered by a health care professional. If your partner is found to be a carrier for 21-Hydroxylase Deficiency, 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 21-Hydroxylase Deficiency
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for 21-Hydroxylase Deficiency
- Adoption or use of a sperm or egg donor who is not a carrier for 21-Hydroxylase Deficiency

What resources are available?

- Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/21-hydroxylase-deficiency>
- GeneReviews: <https://www.ncbi.nlm.nih.gov/books/NBK1171/>
- Prenatal diagnosis by CVS: <http://www.marchofdimes.org/chorionic-villus-sampling.aspx>

Patient Information

Patient Name:

Test Information

Ordering Physician:



Date Of Birth:



Clinic Information:

Case File ID:



Report Date:

- Prenatal diagnosis by amniocentesis: <http://www.marchofdimes.org/amniocentesis.aspx>
- PGD with IVF: <http://www.natera.com/spectrum>

Patient Information

Patient Name: [REDACTED]

Test Information

Ordering Physician: [REDACTED]



Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Clinic Information:

Report Date:

CYSTIC FIBROSIS**Understanding Your Horizon Carrier Screen Results****What are Cystic Fibrosis and CFTR-Related Disorders?**

Cystic Fibrosis (CF) and CFTR-Related Disorders are inherited disorders that affect many different areas of the body including the lungs, digestive system, and fertility. CF and CFTR-Related Disorders do not affect intelligence. Signs and symptoms of CF start in early childhood and include delayed growth caused by problems in digestion and repeated lung infections that lead to permanent lung damage. Children and adults with CF usually have frequent hospitalizations because of lung infections. Over time, complications of CF can lead to lung transplants and early death. CFTR-Related Disorders cause less severe symptoms, and some only affect male fertility. There are treatments for CF that can lessen the severity of the symptoms; however, there is currently no cure. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes CF and CFTR-Related Disorders?

CF and CFTR-Related Disorders are caused by a change, or mutation, in both copies of the CFTR gene pair. These mutations cause the genes to not work properly or not work at all. When both copies of this gene do not work correctly, mucus and other body fluids become thick and sticky. This causes problems with how the lungs, digestive system, and other body systems function and leads to the symptoms described above. CF and CFTR-Related Disorders are inherited in an autosomal recessive manner. This means that, in most cases, both parents must be carriers of a mutation in one copy of the CFTR gene to have a child with CF or CFTR-Related Disorders. People who are CF or CFTR-Related Disorders carriers do not have CF or CFTR-Related Disorders themselves. Usually a child inherits two copies of each gene, one copy from the mother and one copy from the father. If the mother and father are both carriers for CF and CFTR-Related Disorders, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their CFTR gene mutations to the child, who will then have CF or CFTR-Related Disorders. Although most CFTR gene mutations cause classic CF, there are some specific CFTR mutations that cause less severe symptoms, and some only affect male fertility. It is sometimes, but not always, possible to determine whether a specific CFTR mutation causes classic CF or a milder form of the condition. A small number of CF carriers may have mild respiratory or other symptoms. If you have concerns about your own health or symptoms, we encourage you to discuss your results and health history with your health care provider. Individuals found to carry more than one mutation in the CFTR genes should discuss their risk for having an affected child, and any potential effects to their own health, with their health care provider.

What can I do next?

You may wish to speak with a local genetic counselor about your CF and CFTR-Related Disorders carrier test results. A genetic counselor in your region 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 CF and CFTR-Related Disorders ordered by a health care professional. If your partner is not found to be a carrier of CF and CFTR-Related Disorders, your risk of having a child with CF and CFTR-Related Disorders is greatly reduced. Couples at risk of having a baby with CF and CFTR-Related Disorders 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 CF and CFTR-Related Disorders. Although CF is screened for as part of the Newborn Screening program in some US states, babies at 25% for this condition may need diagnostic testing in addition to newborn screening. If you are not yet pregnant, your partner can have CF and CFTR-Related Disorders carrier testing ordered by a health care professional. If your partner is found to be a carrier for CF and CFTR-Related Disorders, 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 CF
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for CF and CFTR-Related Disorders
- Adoption or use of a sperm or egg donor who is not a carrier for CF and CFTR-Related Disorders

What resources are available?

- Cystic Fibrosis Foundation: www.cff.org
- Gene Reviews: Cystic Fibrosis: <https://www.ncbi.nlm.nih.gov/books/NBK1250/>
- 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: [REDACTED]

Test Information

Ordering Physician: [REDACTED]



Clinic Information: [REDACTED]

Date Of Birth: [REDACTED]

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POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE**Understanding Your Horizon Carrier Screen Results****What is Polycystic Kidney Disease, Autosomal Recessive?**

Polycystic Kidney Disease, Autosomal Recessive (ARPKD) is an inherited disorder that affects the kidneys and other organs, including the liver. Affected children are typically born with enlarged kidneys with multiple fluid-filled sacs called cysts. The kidneys do not work properly causing serious health problems. In the most severe form, the kidney problems begin in pregnancy and may affect fetal lung development because of low fluid levels in the pregnancy (oligohydramnios) caused by the fetal kidney disease. Infants born with this severe form of ARPKD often have very serious lung disease that may lead to early death. In some cases, the kidney cysts, along with progressive loss of kidney function do not develop until later infancy or childhood. Liver disease (congenital hepatic fibrosis) occurs in about half of all children with ARPKD. Currently there is no cure for this condition, although medical treatment, which may include kidney and/or liver transplantation, is available. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Polycystic Kidney Disease, Autosomal Recessive?

Polycystic Kidney Disease, Autosomal Recessive (ARPKD) is caused by a gene change, or mutation, in both copies of the PKHD1 gene pair. These mutations cause the genes to not work properly or not work at all. When both copies of this gene do not work correctly, the kidneys do not develop properly and liver disease may also occur, leading to the symptoms described above. ARPKD 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 PKHD1 gene to have a child with ARPKD. People who are carriers for ARPKD are usually healthy and do not have symptoms nor do they have ARPKD 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 ARPKD, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their PKHD1 gene mutations to the child, who will then have the condition. Individuals found to carry more than one mutation for ARPKD should discuss their risk for having an affected child, and any potential effects to their own health, with their health care provider.

What can I do next?

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

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

What resources are available?

- ARPKD CHF Alliance: <http://www.arpkdchf.org/>
- PKD Foundation: <http://www.pkdcure.org/>
- Prenatal diagnosis done through CVS: <http://www.marchofdimes.org/chorionic-villus-sampling.aspx>
- Prenatal diagnosis done through Amniocentesis: <http://www.marchofdimes.org/amniocentesis.aspx>
- Preimplantation genetic diagnosis (PGD) with IVF: <http://www.natera.com/spectrum>

Patient Information

Patient Name: [REDACTED]

Test Information

Ordering Physician: [REDACTED]



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WILSON DISEASE**Understanding Your Horizon Carrier Screen Results****What is Wilson Disease?**

Wilson Disease is an inherited disorder that causes copper from the diet to build up in certain parts of the body, especially in the liver, eyes, and brain. Signs and symptoms of Wilson Disease usually begin in the teenage years and in rare cases not until adulthood. Symptoms include liver disease, nervous system and psychiatric problems, and specific eye findings called Kayser-Fleischer rings (green/brown colored areas of excess copper on the surface of the eyes that do not interfere with vision). Other symptoms may include problems with coordination, movement, and behavior. Wilson Disease is commonly treated through chelation therapy to remove the excess stored copper from the body. This treatment helps to slow, and in some cases stop, the progression of the disease and improve symptoms. With treatment, people with Wilson Disease can have a normal lifespan. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Wilson Disease?

Wilson Disease is caused by a change, or mutation, in both copies of the ATP7B gene pair. These mutations cause the genes to not work properly or not work at all. Normal function of the ATP7B genes is needed for normal transport of copper within the cells of the body. When both copies of the ATP7B gene do not work correctly, it leads to the symptoms described above. Wilson Disease 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 ATP7B gene to have a child with Wilson Disease. People who are carriers for Wilson Disease are usually healthy and do not have symptoms nor do they have Wilson Disease 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 Wilson Disease there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their ATP7B gene mutations to the child, who will then have the condition. Individuals found to carry more than one mutation for Wilson Disease should discuss their risk for having an affected child, and any potential risks to their own health, with their health care provider.

What can I do next?

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

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

What resources are available?

- Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/wilson-disease>
- 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: [REDACTED]

Test Information

Ordering Physician: [REDACTED]



Date Of Birth: [REDACTED]

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VARIANT DETAILS**ATP7B, c.3207C>A (p.H1069Q), heterozygous, pathogenic**

- The c.3207C>A (p.H1069Q) variant in the ATP7B gene has been observed at a frequency of 0.1019% 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 Wilson disease (PMID: 20517649, 26819605).
- This variant has been reported in ClinVar [ID: 3848].

CFTR, c.1652G>A (p.G551D), heterozygous, pathogenic

- The c.1652G>A (p.G551D) variant in the CFTR gene has been observed at a frequency of 0.0181% 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 cystic fibrosis (PMID:).
- This variant has been reported in ClinVar [ID: 7120].

CYP21A2, c.844G>T (p.V282L) [Legacy name: V281L], heterozygous, pathogenic

- The c.844G>T (p.V282L) [Legacy name: V281L] variant in the CYP21A2 gene has been observed at a frequency of 0.5515% 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 non-classic congenital adrenal hyperplasia (PMID: 19263525, 25041270, 32616876).
- Functional studies demonstrated that this variant causes reduced enzyme activity (PMID: 24953648).
- This variant has been reported in ClinVar [ID: 12151].

PKHD1, c.9107T>G (p.V3036G), heterozygous, likely pathogenic

- The c.9107T>G (p.V3036G) variant in the PKHD1 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 polycystic kidney disease (PMID: 27225849, 15805161).
- This variant has been reported in ClinVar [ID: 594333].

Patient Information

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DISEASES SCREENED

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

Autosomal Recessive

1

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

3

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

5

5-ALPHA-REDUCTASE DEFICIENCY (*SRD5A2*) **negative**

6

6-PYRUVYL-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-GOUTIERES SYNDROME (*SAMHD1*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2A-RELATED (*RNASEH2A*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2B-RELATED (*RNASEH2B*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2C-RELATED (*RNASEH2C*) **negative**
AICARDI-GOUTIERES SYNDROME, TREX1-RELATED (*TREX1*) **negative**
ALPHA-MANNOSIDOSIS (*MAN2B1*) **negative**
ALPHA-THALASSEMIA (*HBA1/HBA2*) **negative**
ALPORT SYNDROME, COL4A3-RELATED (*COL4A3*) **negative**
ALPORT SYNDROME, COL4A4-RELATED (*COL4A4*) **negative**
ALSTROM SYNDROME (*ALMS1*) **negative**
AMISH INFANTILE EPILEPSY SYNDROME (*ST3GAL5*) **negative**
ANDERMANN SYNDROME (*SLC12A6*) **negative**
ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (*GATM*) **negative**
ARGININEMIA (*ARG1*) **negative**
ARGININOSUCCINATE LYASE DEFICIENCY (*ASL*) **negative**
AROMATASE DEFICIENCY (*CYP19A1*) **negative**
ASPARAGINE SYNTHETASE DEFICIENCY (*ASNS*) **negative**
ASPARTYLGLYCOSAMINURIA (AGA) **negative**
ATAXIA WITH VITAMIN E DEFICIENCY (*TTPA*) **negative**
ATAXIA-TELANGIECTASIA (*ATM*) **negative**
ATAXIA-TELANGIECTASIA-LIKE DISORDER 1 (*MRE11*) **negative**
ATRANSFERRINEMIA (*TF*) **negative**
AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (*SLC35A3*) **negative**
AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (*AIRE*) **negative**
AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (ARCI), SLC27A4-RELATED (*SLC27A4*) **negative**
AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (SACS) **negative**

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 FRONTOPIRIETAL POLYMICROGYRIA (*GPR56*) **negative**

BIOTINIDASE DEFICIENCY (*BTD*) **negative**

BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBGD) (*SLC19A3*) **negative**

BLOOM SYNDROME (*BLM*) **negative**

BRITTLE CORNEA SYNDROME 1 (*ZNF469*) **negative**

BRITTLE CORNEA SYNDROME 2 (*PRDM5*) **negative**

C

CANAVAN DISEASE (*ASPA*) **negative**
CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (*CPS1*) **negative**
CARNITINE DEFICIENCY (*SLC22A5*) **negative**
CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY (*CPT1A*) **negative**
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (*CPT2*) **negative**
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (*SLC25A20*) **negative**
CARPENTER SYNDROME (*RAB23*) **negative**
CARILAGE-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 (*ACS3F3*) **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*) **see first page**
CONGENITAL ADRENAL INSUFFICIENCY, CYP11A1-RELATED (*CYP11A1*) **negative**
CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (*MPL*) **negative**
CONGENITAL CHRONIC DIARRHEA (*DGAT1*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, ALG1-RELATED (*ALG1*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-Related (*PMM2*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (*MPL*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (*ALG6*) **negative**
CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (*SEC23B*) **negative**
CONGENITAL FINNISH NEPHROSIS (*NPHS1*) **negative**
CONGENITAL HYDROCEPHALUS 1 (*CCDC88C*) **negative**
CONGENITAL HYPERINSULINISM, KCNJ11-Related (*KCNJ11*) **negative**
CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (CIPA) (*NTRK1*) **negative**
CONGENITAL MYASTHENIC SYNDROME, CHAT-RELATED (*CHAT*) **negative**
CONGENITAL MYASTHENIC SYNDROME, CHRNE-RELATED (*CHRNE*) **negative**
CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED (*COLQ*) **negative**
CONGENITAL MYASTHENIC SYNDROME, DOK7-RELATED (*DOK7*) **negative**
CONGENITAL MYASTHENIC SYNDROME, RAPSIN-RELATED (*RAPSIN*) **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*) **see first page**
CYSTINOSIS (*CTNS*) **negative**
CYTOCHROME C OXIDASE DEFICIENCY, PET100-RELATED (*PET100*) **negative**
CYTOCHROME P450 OXIOREDUCTASE DEFICIENCY (*POR*) **negative**

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D

D-BIFUNCTIONAL PROTEIN DEFICIENCY (*HSD17B4*) **negative**
DEAFNESS, AUTOSOMAL RECESSIVE 77 (*LOXHD1*) **negative**
DIHYDROPTERIDINE REDUCTASE (DHPR) DEFICIENCY (*QDPR*) **negative**
DONNAI-BARROW SYNDROME (*LRP2*) **negative**
DUBIN-JOHNSON SYNDROME (*ABCC2*) **negative**
DYSKERATOSIS CONGENITA SPECTRUM DISORDERS (*TERT*) **negative**
DYSKERATOSIS CONGENITA, RTKL1-RELATED (*RTKL1*) **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**
ISOLEUCIC 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**

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**L**

LAMELLAR ICHTHYOSIS, TYPE 1 (*TGM1*) **negative**
LARON SYNDROME (*GHR*) **negative**
LEBER CONGENITAL AMAUROSIS 2 (*RPE65*) **negative**
LEBER CONGENITAL AMAUROSIS TYPE APL1 (*APL1*) **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 CBLF (*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) (*AGLU*) **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-ACETYLGUTAMATE SYNTHASE DEFICIENCY (*NAGS*) **negative**
NEMALINE MYOPATHY, NEB-RELATED (*NEB*) **negative**
NEPHRONOPHTHISIS 1 (*NPHP1*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, CLN5-RELATED (*CLN5*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, CLN6-RELATED (*CLN6*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, CLN8-RELATED (*CLN8*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, MFSD8-RELATED (*MFSD8*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, PPT1-RELATED (*PPT1*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, TPP1-RELATED (*TPP1*) **negative**
NGLY1-CONGENITAL DISORDER OF GLYCOSYLATION (*NGLY1*) **negative**
NIEMANN-PICK DISEASE, TYPE C1 / D (*NPC1*) **negative**
NIEMANN-PICK DISEASE, TYPE C2 (*NPC2*) **negative**
NIEMANN-PICK DISEASE, TYPES A / B (*SMPD1*) **negative**
NIJMEGEN BREAKAGE SYNDROME (*NBN*) **negative**
NON-SYNDROMIC HEARING LOSS, GJB2-RELATED (*GJB2*) **negative**
NON-SYNDROMIC HEARING LOSS, MYO15A-RELATED (*MYO15A*) **negative**
NONSYNDROMIC HEARING LOSS, OTOA-RELATED (*OTOA*) **negative**
NONSNDROMIC HEARING LOSS, OTOF-RELATED (*OTOF*) **negative**
NONSNDROMIC HEARING LOSS, PJVK-RELATED (*PJVK*) **negative**
NONSNDROMIC HEARING LOSS, SYNE4-RELATED (*SYNE4*) **negative**
NONSNDROMIC HEARING LOSS, TMC1-RELATED (*TMC1*) **negative**
NONSNDROMIC HEARING LOSS, TMRSS3-RELATED (*TMRSS3*) **negative**
NONSNDROMIC INTELLECTUAL DISABILITY (*CC2D1A*) **negative**
NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (*SAMD9*) **negative**

O

OCULOCUTANEOUS ALBINISM TYPE IV (*SLC45A2*) **negative**
OCULOCUTANEOUS ALBINISM TYPE, III (*TYRP1*) **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**
PAILLON LÉFÈVRE 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**

Patient Information

Patient Name:

Test Information

Ordering Physician:



Clinic Information:

Date Of Birth:



Case File ID:



Report Date:

P

POLG-RELATED DISORDERS (POLG) **negative**
POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE (PKHD1) **see first page**
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, 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 (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 (TBCE) **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**
PYCNODYSTOSIS (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**
RYYR1-RELATED CONDITIONS (RYYR1) **negative**

S

SALLA DISEASE (SLC17A5) **negative**
SANDHOFF DISEASE (HEXB) **negative**
SCHIMKE IMMUNOOSESQU DYSPLASIA (SMARCA1) **negative**
SCHINDLER DISEASE (NAGA) **negative**
SEGAWA SYNDROME, TH-RELATED (TH) **negative**
SENIOR-LOKEN SYNDROME 4/NEPHRONOPHTHISIS 4 (NPHP4) **negative**
SEPIAPTERIN REDUCTASE DEFICIENCY (SPR) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3D-RELATED (CD3D) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3E-RELATED (CD3E) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), FOXP1-RELATED (FOXP1) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), IKKB-RELATED (IKKB) **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 POLYDACTYL (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 (VWOOX) **negative**
SPONDYLOCOSTAL DYSOSTOSIS 1 (DLL3) **negative**
SPONDYLOTHORACIC DYSOSTOSIS, MESP2-Related (MESP2) **negative**
STEEL SYNDROME (COL27A1) **negative**
STERIOD-RESISTANT NEPHROTIC SYNDROME (NPHS2) **negative**
STUVE-WIEDEMANN SYNDROME (LIFR) **negative**
SURF1-RELATED CONDITIONS (SURF1) **negative**
SURFACTANT DYSFUNCTION, ABCA3-RELATED (ABCA3) **negative**

T

TAY-SACHS DISEASE (HEXA) **negative**
TBCE-RELATED CONDITIONS (TBCE) **negative**
THIAMINE-RESPONSIVE MEGALOBlastic ANEMIA SYNDROME (SLC19A2) **negative**
THYROID DYSHORMONOGENESIS 1 (SLC5A5) **negative**
THYROID DYSHORMONOGENESIS 2A (TPO) **negative**
THYROID DYSHORMONOGENESIS 3 (TG) **negative**
THYROID DYSHORMONOGENESIS 6 (DUOX2) **negative**
TRANSCOBALAMIN II DEFICIENCY (TCN2) **negative**
TRICHOHEPATOENTERIC SYNDROME, SKIC2-RELATED (SKIC2) **negative**
TRICHOHEPATOENTERIC SYNDROME, TTC37-RELATED (TTC37) **negative**
TRICHOHYDROSTROPHY 1/XERODERMA PIGMENTOSUM, GROUP D (ERCC2) **negative**
TRIMETHYLAMINURIA (FMO3) **negative**
TRIPLE A SYNDROME (AAA5) **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) **see first page**
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**

Patient Information

Patient Name:

Test Information

Ordering Physician:



Date Of Birth:



Clinic Information:

Case File ID:



Report Date:

Z

ZELLWEGER SPECTRUM DISORDERS, PEX2-RELATED (PEX2) **negative**
ZELLWEGER SPECTRUM DISORDERS, PEX6-RELATED (PEX6) **negative**

Patient Information

Patient Name:

Test Information

Ordering Physician:



Date Of Birth:



Case File ID:



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 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, 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 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

Patient Name: [REDACTED]

Test Information

Ordering Physician: [REDACTED]



Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Clinic Information:

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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.

7428,DONOR ▲

DOB: [REDACTED] Age: [REDACTED] Specimen: [REDACTED]
 Sex: M Fasting: [REDACTED] Requisition: [REDACTED]
 Phone: [REDACTED] Lab Reference: [REDACTED]
 Patient ID: 7428 [REDACTED] Report Status: FINAL / SEE REPORT

Collected: 07/02/2024 07:30
 Received: 07/03/2024 17:02
 Reported: 07/12/2024 11:52

[REDACTED]
 [REDACTED]
 [REDACTED]
 [REDACTED]
 [REDACTED]
 [REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

Hemoglobinopathy Evaluation

(FINAL)

Lab: AMD

Hemoglobinopathy Evaluation

No Historical Data

(FINAL)

Red Blood Cell Count

Reference Range: 4.20-5.80 Mill/uL

5.34

(FINAL)



No Historical Data

HEMOGLOBIN

Reference Range: 13.2-17.1 g/dL

15.7

(FINAL)



No Historical Data

Hematocrit

No Historical Data

(FINAL)

Hematocrit

Reference Range: 38.5-50.0 %

47.9

(FINAL)



No Historical Data

MCV

Reference Range: 80.0-100.0 fL

89.7

(FINAL)



No Historical Data

MCH

Reference Range: 27.0-33.0 pg

29.4

(FINAL)



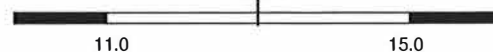
No Historical Data

RDW

Reference Range: 11.0-15.0 %

13.0

(FINAL)



No Historical Data

Hemoglobinopathy Evaluation

No Historical Data

(FINAL)

Hemoglobin A

Reference Range: >96.0 %

97.4

(FINAL)



No Historical Data

Hemoglobin F

Reference Range: <2.0 %

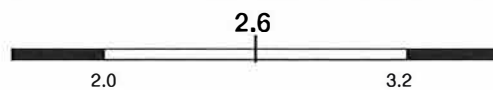


FINAL

No Historical Data

Hemoglobin A2 (Quant)

Reference Range: 2.0-3.2 %



FINAL

No Historical Data

Interpretation

No Historical Data

FINAL

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

FINAL

Chromosome Analysis, Blood

No Historical Data

FINAL

Order ID: [REDACTED]
Specimen Type: Blood
Clinical Indication: Genetic 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.

Benjamin A, Hilton, Ph.D., FACMG, Technical Director, Cytogenetics and Genomics, 703-802-7156

Electronic Signature: 7/12/2024 11:07 AM

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

Note: Data displayed only for results that meet strict identification matching. Historical result view may vary based on corrected or updated patient demographics. The reference range