Fairfax Cryobank

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

SPERM DONOR GENETIC TESTING SUMMARY

Donor # 7176

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

Last Updated: 04/15/2025

Donor Reported Ancestry: Puerto Rican

Jewish Ancestry: No

Genetic Test*	**Result**	**Comments** **Donor's Residual Risk****
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Expanded Genetic Disease Carrier Screening Panel attached - 549 diseases by gene sequencing and del/dup analysis.	Carrier: Congenital Adrenal Hyperplasia, 21 - Hydroxylase Deficiency (CYP21A2) Carrier: Megalencephalic Leukoencephalopathy With Subcortical Cysts (MLC1) Carrier: Mucopolysaccharidosis, Type Iv B/Gm1 Gangliosidosis (GLB1) Negative for other genes tested.	Partner testing is recommended before using this donor.

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

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

Patient Information	
Patient Name:	Donor 7176
Date Of Birth:	[redacted]
Gender:	Male
Ethnicity:	Hispanic/Latin American
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:	04/01/2025
Sample Collected:	03/19/2025
Sample Received:	03/20/2025
Sample Type:	Blood

horizon™ natera carrier screen

CARRIER SCREENING REPORT

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

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

FINAL RESULTS SUMMARY:

CARRIER for Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficiency

Positive for the pathogenic variant c.92C>T (p.P31L) [Legacy name: P30L] 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 or simple virilizing congenital adrenal hyperplasia (PMID: 23359698, 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 Megalencephalic Leukoencephalopathy With Subcortical Cysts

Positive for the pathogenic variant partial exon 4 and exon 5 deletion in the MLC1 gene. If this individual's partner is a carrier for MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS, 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 Mucopolysaccharidosis, Type Iv B/Gm1 Gangliosidosis

Positive for the pathogenic variant c.1258A>C (p.T420P) in the GLB1 gene. If this individual's partner is a carrier for MUCOPOLYSACCHARIDOSIS, TYPE IV B/GM1 GANGLIOSIDOSIS, 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 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 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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Laboratory Director, Baylor Genetics

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Laboratory Director, Natera

natera® Conceive. Deliver.

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 21Hydroxylase 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:

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

Test Information
Ordering Physician: [redacted]

Clinic Information:

Report Date:

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

Patient Information
Patient Name:

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

Test Information
Ordering Physician: [redacted]

Clinic Information:

Report Date:

MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS

Understanding Your Horizon Carrier Screen Results

What is Megalencephalic Leukoencephalopathy with Subcortical Cysts?

Megalencephalic Leukoencephalopathy with Subcortical Cysts (Type 1) is an inherited disorder that affects the brain and nervous system. Signs and symptoms begin in infancy or childhood and include large head and brain size, developmental delays, loss of developmental skills, problems with coordination and movement, muscle stiffness, seizures, speech problems, and mild to moderate intellectual disability. Some people with this condition can walk without assistance and others eventually need a wheelchair. Currently there is no cure for this condition and treatment is based on symptoms. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Megalencephalic Leukoencephalopathy with Subcortical Cysts?

Megalencephalic Leukoencephalopathy with Subcortical Cysts is caused by gene changes, or mutations, in one of several gene pairs including the MLC1 gene pair. These mutations cause the genes to not work properly or not work at all. Normal function of the MLC1 genes is important for development of the brain and nerves. When both copies of the MLC1 gene pair do not work correctly, it leads to the symptoms described above. Megalencephalic Leukoencephalopathy with Subcortical Cysts (Type 1) is inherited in an autosomal recessive manner. This means that, in most cases, both parents must be carriers of a mutation in one copy of the MLC1 gene to have a child with Megalencephalic Leukoencephalopathy with Subcortical Cysts. People who are carriers for Megalencephalic Leukoencephalopathy with Subcortical Cysts (Type 1) 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 Megalencephalic Leukoencephalopathy with Subcortical Cysts, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their MLC1 gene mutations to the child, who will then have this condition. Individuals found to carry more than one mutation for Megalencephalic Leukoencephalopathy with Subcortical Cysts should discuss their risk for having an affected child with their health care provider. There are other forms of Megalencephalic Leukoencephalopathy with Subcortical Cysts, called Types 2A and B, both caused by mutations in a different gene. People who are carriers for a mutation in the MLC1 gene are not likely to be at increased risk for having children with these other forms of the condition.

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 Megalencephalic Leukoencephalopathy with Subcortical Cysts ordered by a health care professional. If your partner is not found to be a carrier for Megalencephalic Leukoencephalopathy with Subcortical Cysts, your risk of having an affected child is greatly reduced. Couples at risk of having a baby with Megalencephalic Leukoencephalopathy with Subcortical Cysts 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 Megalencephalic Leukoencephalopathy with Subcortical Cysts ordered by a health care professional. If your partner is found to be a carrier for Megalencephalic Leukoencephalopathy with Subcortical Cysts, you have several reproductive options to consider:

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

What resources are available?

- Genetics Home Reference: http://ghr.nlm.nih.gov/condition/megalencephalic- leukoencephalopathy-with-subcortical-cysts
- GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1535/
- 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:

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

Test Information
Ordering Physician: [redacted]

Clinic Information:

Report Date:

horizon™ natera carrier screen

MUCOPOLYSACCHARIDOSIS, TYPE IV B/GM1 GANGLIOSIDOSIS

Understanding Your Horizon Carrier Screen Results

What is Mucopolysaccharidosis, Type IVB/GM1 Gangliosidosis?

Mucopolysaccharidosis (MPS), Type IVB (also called Morquio Syndrome) and GM1 Gangliosidosis are inherited disorders that affect many parts of the body. Both disorders are caused by mutations in the same gene but they have different signs and symptoms. The more common disorder, GM1 Gangliosidosis, causes progressive loss of nerve cells in the brain and spine. The infantile form of GM1 Gangliosidosis causes weakened muscles, loss of motor skills, developmental delay and intellectual disability, clouding of the cornea of the eye and degeneration of the retina that causes vision loss, and enlargement of the liver, spleen and heart. Babies with this form usually die by early childhood. Some children with GM1 Gangliosidosis do not start showing symptoms until early childhood and do not have organ enlargement but still have loss of skills and a shortened lifespan. In rare cases symptoms do not start until the teenage or early adult years and include episodes of muscle spasms (dystonia), problems with walking and speech, enlarged heart, and memory loss; this adult-onset form is mostly seen in people of Japanese ancestry. The less common disorder, MPS, Type IVB, causes skeletal abnormalities, and abnormal growth of bone and cartilage. Other signs and symptoms of MPS, Type IVB often include short stature, overly mobile joints, hearing loss, breathing problems, spinal cord problems, hernias, sleep apnea, heart disease, multiple cavities, and clouding of the cornea of the eye. Intelligence is not affected. Lifespan is decreased in children with the early-onset form of MPS, Type IVB with death often occurring in late childhood or early teens. Lifespan may be near normal in people with the later-onset form. In some cases, affected individuals have been treated with or participated in clinical trials using stem cell transplantation from cord blood or bone marrow. Couples at risk of having an affected child may consider cord blood banking, as siblings have a higher chance of being a match for stem cell transplantation than a non-related individual. More information can be found at: https://parentsguidecordblood.org/en. Clinical trials involving potential new treatments for these conditions may be available (see www.clinicaltrials.gov). It is sometimes, but not always, possible to tell whether a specific gene mutation will cause GM1 Gangliosidosis or MPS, Type IVB. As discussed below, they are both inherited in the same manner and have the same reproductive options available.

What causes Mucopolysaccharidosis, Type IVB/GM1 Gangliosidosis?

MPS, Type IVB and GM1 Gangliosidosis are each caused by a change, or mutation, in both copies of the GLB1 gene pair, which cause the genes to not work properly or not work at all. When both copies of the GLB1 gene do not work properly, it leads to the symptoms of either GM1 Gangliosidosis or MPS, Type IVB as described above. MPS, Type IVB and GM1 Gangliosidosis are both 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 GLB1 gene to have a child with MPS, Type IVB or GM1 Gangliosidosis. People who are carriers for MPS, Type IVB or GM1 Gangliosidosis are usually healthy and do not have symptoms nor do they have either condition 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 MPS, Type IVB or GM1 Gangliosidosis there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their GLB1 gene mutations to the child, who will then have one of these conditions. Individuals found to carry more than one mutation for MPS, Type IVB or GM1 Gangliosidosis should discuss their risk for having an affected child and any potential effects to their own health with their health care provider. There are many other types of Mucopolysaccharidosis (MPS) and Gangliosidosis, each caused by mutations in different genes. A carrier for MPS, Type IVB or GM1 Gangliosidosis is not likely to be at increased risk for having children with the other forms of MPS or Gangliosidosis.

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 MPS, Type IVB and GM1 Gangliosidosis ordered by a health care professional. If your partner is not found to be a carrier for either disorder, your risk of having a child with either condition is greatly reduced. Couples at risk of having a baby with MPS, Type IVB or GM1 Gangliosidosis 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 these conditions. If you are not yet pregnant, your partner can have carrier screening for MPS, Type IVB and GM1 Gangliosidosis ordered by a health care professional. If your partner is found to be a carrier for either disorder, you have several reproductive options to consider:

- Natural pregnancy with or without prenatal diagnosis of the fetus or testing the baby after birth for MPS, Type IVB or GM1 Gangliosidosis
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for MPS, Type IVB or GM1 Gangliosidosis
- Adoption or use of a sperm or egg donor who is not a carrier for MPS, Type IVB or GM1 Gangliosidosis

What resources are available?

- Genetics Home Reference: http://ghr.nlm.nih.gov/condition/gm1-gangliosidosis
- Genetics Home Reference: http://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-iv
- 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

VARIANT DETAILS

CYP21A2, c.92C>T (p.P31L) [Legacy name: P30L], pathogenic

- The c.92C>T (p.P31L) [Legacy name: P30L] variant in the CYP21A2 gene has been observed at a frequency of 0.0158% 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 or simple virilizing congenital adrenal hyperplasia (PMID: 23359698, 25041270, 32616876).
- Functional studies demonstrated that this variant causes reduced enzyme activity (PMID: 24953648).
- This variant has been reported in ClinVar [ID: 12153].

GLB1, c.1258A>C (p.T420P), pathogenic

- The c.1258A>C (p.T420P) variant in the GLB1 gene has not been observed in the gnomAD v2.1.1 dataset.
- This variant has been reported in conjunction with another variant in individuals with GM1-gangliosidosis (PMID: 16941474, 17664528).
- Functional studies have demonstrated this variant causes loss of enzymatic activity (PMID: 16941474, 17664528).
- This variant has been described in ClinVar [ID: 1070505].

MLC1, partial exon 4 and exon 5 deletion, pathogenic

- The partial exon 4 and exon 5 deletion in the MLC1 gene is predicted to be in-frame in a gene where loss-of-function is a known mechanism of disease. It impacts a significant portion of the protein length or a critical region of the protein, potentially disrupting normal protein function.
- Copy number loss involving this region has been reported in a homozygous state in individual(s) with megalencephalic leukoencephalopathy with subcortical cysts (PMID: 11935341, 16652334).
- This variant has been described in ClinVar (ID: 660628).

Patient Information
Patient Name:

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

Test Information
Ordering Physician: [redacted]

Clinic Information:

Report Date:

horizon™ natera carrier screen

DISEASES SCREENED

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

Autosomal Recessive

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

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

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

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

A
ABCA4-RELATED CONDITIONS (*ABCA4*) **negative**
ABETALIPOPROTEINEMIA (*MTTP*) **negative**
ACHONDROGENESIS, TYPE 1B (*SLC26A2*) **negative**
ACHROMATOPSIA, CNGB3-RELATED (*CNGB3*) **negative**
ACRODERMATITIS ENTEROPATHICA (*SLC39A4*) **negative**
ACTION MYOCLONUS-RENAL FAILURE (AMRF) SYNDROME (*SCARB2*) **negative**
ACUTE INFANTILE LIVER FAILURE, TRMU-RELATED (*TRMU*) **negative**
ACYL-COA OXIDASE I DEFICIENCY (*ACOX1*) **negative**
AICARDI-GOUTIÈRES SYNDROME (*SAMHD1*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2A-RELATED (*RNASEH2A*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2B-RELATED (*RNASEH2B*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2C-RELATED (*RNASEH2C*) **negative**
AICARDI-GOUTIÈRES SYNDROME, TREX1-RELATED (*TREX1*) **negative**
ALPHA-MANNOSIDOSIS (*MAN2B1*) **negative**
ALPHA-THALASSEMIA (*HBA1/HBA2*) **negative**
ALPORT SYNDROME, COL4A3-RELATED (*COL4A3*) **negative**
ALPORT SYNDROME, COL4A4-RELATED (*COL4A4*) **negative**
ALSTROM SYNDROME (*ALMS1*) **negative**
AMISH INFANTILE EPILEPSY SYNDROME (*ST3GAL5*) **negative**
ANDERMANN SYNDROME (*SLC12A6*) **negative**
ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (*GATM*) **negative**
ARGININEMIA (*ARG1*) **negative**
ARGININOSUCCINATE LYASE DEFICIENCY (*ASL*) **negative**
AROMATASE DEFICIENCY (*CYP19A1*) **negative**
ASPARAGINE SYNTHETASE DEFICIENCY (*ASNS*) **negative**
ASPARTYLGLYCOSAMINURIA (*AGA*) **negative**
ATAXIA WITH VITAMIN E DEFICIENCY (*TTPA*) **negative**
ATAXIA-TELANGIECTASIA (*ATM*) **negative**
ATAXIA-TELANGIECTASIA-LIKE DISORDER 1 (*MRE11*) **negative**
ATRANSFERRINEMIA (*TF*) **negative**
AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (*SLC35A3*) **negative**
AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (*AIRE*) **negative**
AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (ARCI), SLC27A4-RELATED (*SLC27A4*) **negative**
AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (*SACS*) **negative**

B
BARDET-BIEDL SYNDROME, ARL6-RELATED (*ARL6*) **negative**
BARDET-BIEDL SYNDROME, BBS10-RELATED (*BBS10*) **negative**
BARDET-BIEDL SYNDROME, BBS12-RELATED (*BBS12*) **negative**
BARDET-BIEDL SYNDROME, BBS1-RELATED (*BBS1*) **negative**
BARDET-BIEDL SYNDROME, BBS2-RELATED (*BBS2*) **negative**
BARDET-BIEDL SYNDROME, BBS4-RELATED (*BBS4*) **negative**
BARDET-BIEDL SYNDROME, BBS5-RELATED (*BBS5*) **negative**
BARDET-BIEDL SYNDROME, BBS7-RELATED (*BBS7*) **negative**
BARDET-BIEDL SYNDROME, BBS9-RELATED (*BBS9*) **negative**
BARDET-BIEDL SYNDROME, TTC8-RELATED (*TTC8*) **negative**
BARE LYMPHOCYTE SYNDROME, CIITA-RELATED (*CIITA*) **negative**
BARTTER SYNDROME, BSND-RELATED (*BSND*) **negative**
BARTTER SYNDROME, KCNJ1-RELATED (*KCNJ1*) **negative**
BARTTER SYNDROME, SLC12A1-RELATED (*SLC12A1*) **negative**
BATTEN DISEASE, CLN3-RELATED (*CLN3*) **negative**
BETA-HEMOGLOBINOPATHIES (*HBB*) **negative**
BETA-KETOTHIOLASE DEFICIENCY (*ACAT1*) **negative**
BETA-MANNOSIDOSIS (*MANBA*) **negative**
BETA-UREIDOPROPIONASE DEFICIENCY (*UPB1*) **negative**
BILATERAL FRONTOPARIETAL POLYMICROGYRIA (*GPR56*) **negative**
BIOTINIDASE DEFICIENCY (*BTD*) **negative**
BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBGD) (*SLC19A3*) **negative**
BLOOM SYNDROME (*BLM*) **negative**
BRITTLE CORNEA SYNDROME 1 (*ZNF469*) **negative**
BRITTLE CORNEA SYNDROME 2 (*PRDM5*) **negative**

C
CANAVAN DISEASE (*ASPA*) **negative**
CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (*CPS1*) **negative**
CARNITINE DEFICIENCY (*SLC22A5*) **negative**
CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY (*CPT1A*) **negative**
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (*CPT2*) **negative**
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (*SLC25A20*) **negative**
CARPENTER SYNDROME (*RAB23*) **negative**
CARTILAGE-HAIR HYPOPLASIA (*RMRP*) **negative**
CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (*CASQ2*) **negative**
CD59-MEDIATED HEMOLYTIC ANEMIA (*CD59*) **negative**
CEP152-RELATED MICROCEPHALY (*CEP152*) **negative**
CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA (CEDNIK) SYNDROME (*SNAP29*) **negative**
CEREBROTENDINOUS XANTHOMATOSIS (*CYP27A1*) **negative**
CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE C (*PLEKHG5*) **negative**
CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (*NDRG1*) **negative**
CHEDIAK-HIGASHI SYNDROME (*LYST*) **negative**
CHOREOACANTHOCYTOSIS (*VPS13A*) **negative**
CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (*CYBA*) **negative**
CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (*NCF2*) **negative**
CILIOPATHIES, RPGRIP1L-RELATED (*RPGRIP1L*) **negative**
CITRIN DEFICIENCY (*SLC25A13*) **negative**
CITRULLINEMIA, TYPE 1 (*ASS1*) **negative**
CLN10 DISEASE (*CTSD*) **negative**
COHEN SYNDROME (*VPS13B*) **negative**
COL11A2-RELATED CONDITIONS (*COL11A2*) **negative**
COMBINED MALONIC AND METHYLMALONIC ACIDURIA (*ACSF3*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (*GFM1*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (*TSFM*) **negative**
COMBINED PITUITARY HORMONE DEFICIENCY 1 (*POU1F1*) **negative**
COMBINED PITUITARY HORMONE DEFICIENCY-2 (*PROP1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (*CYP11B1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (*CYP17A1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (*CYP21A2*) **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 (*MPI*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (*ALG6*) **negative**
CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (*SEC23B*) **negative**
CONGENITAL FINNISH NEPHROSIS (*NPHS1*) **negative**
CONGENITAL HYDROCEPHALUS 1 (*CCDC88C*) **negative**
CONGENITAL HYPERINSULINISM, KCNJ11-Related (*KCNJ11*) **negative**
CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (CIPA) (*NTRK1*) **negative**
CONGENITAL MYASTHENIC SYNDROME, CHAT-RELATED (*CHAT*) **negative**
CONGENITAL MYASTHENIC SYNDROME, CHRNE-RELATED (*CHRNE*) **negative**
CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED (*COLQ*) **negative**
CONGENITAL MYASTHENIC SYNDROME, DOK7-RELATED (*DOK7*) **negative**
CONGENITAL MYASTHENIC SYNDROME, RAPSN-RELATED (*RAPSN*) **negative**
CONGENITAL NEPHROTIC SYNDROME, PLCE1-RELATED (*PLCE1*) **negative**
CONGENITAL NEUTROPENIA, G6PC3-RELATED (*G6PC3*) **negative**
CONGENITAL NEUTROPENIA, HAX1-RELATED (*HAX1*) **negative**
CONGENITAL NEUTROPENIA, VPS45-RELATED (*VPS45*) **negative**
CONGENITAL SECRETORY CHLORIDE DIARRHEA 1 (*SLC26A3*) **negative**
CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS (*SLC4A11*) **negative**
CORTICOSTERONE METHYLOXIDASE DEFICIENCY (*CYP11B2*) **negative**
COSTEFF SYNDROME (3-METHYLGLUTACONIC ACIDURIA, TYPE 3) (*OPA3*) **negative**
CRB1-RELATED RETINAL DYSTROPHIES (*CRB1*) **negative**
CYSTIC FIBROSIS (*CFTR*) **negative**
CYSTINOSIS (*CTNS*) **negative**
CYTOCHROME C OXIDASE DEFICIENCY, PET100-RELATED (*PET100*) **negative**
CYTOCHROME P450 OXIDOREDUCTASE DEFICIENCY (*POR*) **negative**

D
D-BIFUNCTIONAL PROTEIN DEFICIENCY (*HSD17B4*) **negative**
DEAFNESS, AUTOSOMAL RECESSIVE 77 (*LOXHD1*) **negative**
DIHYDROPTERIDINE REDUCTASE (DHPR) DEFICIENCY (*QDPR*) **negative**
DONNAI-BARROW SYNDROME (*LRP2*) **negative**
DUBIN-JOHNSON SYNDROME (*ABCC2*) **negative**
DYSKERATOSIS CONGENITA SPECTRUM DISORDERS (*TERT*) **negative**
DYSKERATOSIS CONGENITA, RTEL1-RELATED (*RTEL1*) **negative**
DYSTROPHIC EPIDERMOLYSIS BULLOSA, COL7A1-Related (*COL7A1*) **negative**

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

F
FACTOR XI DEFICIENCY (*F11*) **negative**
FAMILIAL DYSAUTONOMIA (*IKBKAP*) **negative**
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, PRF1-RELATED (*PRF1*) **negative**
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STX11-RELATED (*STX11*) **negative**
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STXBP2-RELATED (*STXBP2*) **negative**
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, UNC13D-RELATED (*UNC13D*) **negative**
FAMILIAL HYPERCHOLESTEROLEMIA, LDLRAP1-RELATED (*LDLRAP1*) **negative**
FAMILIAL HYPERCHOLESTEROLEMIA, LDLR-RELATED (*LDLR*) **negative**
FAMILIAL HYPERINSULINISM, ABCC8-RELATED (*ABCC8*) **negative**
FAMILIAL NEPHROGENIC DIABETES INSIPIDUS, AQP2-RELATED (*AQP2*) **negative**
FANCONI ANEMIA, GROUP A (*FANCA*) **negative**
FANCONI ANEMIA, GROUP C (*FANCC*) **negative**
FANCONI ANEMIA, GROUP D2 (*FANCD2*) **negative**
FANCONI ANEMIA, GROUP E (*FANCE*) **negative**
FANCONI ANEMIA, GROUP F (*FANCF*) **negative**
FANCONI ANEMIA, GROUP G (*FANCG*) **negative**
FANCONI ANEMIA, GROUP I (*FANCI*) **negative**
FANCONI ANEMIA, GROUP J (*BRIP1*) **negative**
FANCONI ANEMIA, GROUP L (*FANCL*) **negative**
FARBER LIPOGRANULOMATOSIS (*ASAH1*) **negative**
FOVEAL HYPOPLASIA (*SLC38A8*) **negative**
FRASER SYNDROME 3, GRIP1-RELATED (*GRIP1*) **negative**
FRASER SYNDROME, FRAS1-RELATED (*FRAS1*) **negative**
FRASER SYNDROME, FREM2-RELATED (*FREM2*) **negative**
FRIEDREICH ATAXIA (*FXN*) **negative**
FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY (*FBP1*) **negative**
FUCOSIDOSIS, FUCA1-RELATED (*FUCA1*) **negative**
FUMARASE DEFICIENCY (*FH*) **negative**

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

H
HARLEQUIN ICHTHYOSIS (*ABCA12*) **negative**
HEME OXYGENASE 1 DEFICIENCY (*HMOX1*) **negative**
HEMOCHROMATOSIS TYPE 2A (*HFE2*) **negative**
HEMOCHROMATOSIS, TYPE 3, TFR2-Related (*TFR2*) **negative**
HEPATOCEREBRAL MITOCHONDRIAL DNA DEPLETION SYNDROME, MPV17-RELATED (*MPV17*) **negative**
HEREDITARY FRUCTOSE INTOLERANCE (*ALDOB*) **negative**
HEREDITARY HEMOCHROMATOSIS TYPE 2B (*HAMP*) **negative**
HEREDITARY SPASTIC PARAPARESIS, TYPE 49 (*TECPR2*) **negative**
HEREDITARY SPASTIC PARAPLEGIA, CYP7B1-RELATED (*CYP7B1*) **negative**
HERMANSKY-PUDLAK SYNDROME, AP3B1-RELATED (*AP3B1*) **negative**
HERMANSKY-PUDLAK SYNDROME, BLOC1S3-RELATED (*BLOC1S3*) **negative**
HERMANSKY-PUDLAK SYNDROME, BLOC1S6-RELATED (*BLOC1S6*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED (*HPS1*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS3-RELATED (*HPS3*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS4-RELATED (*HPS4*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS5-RELATED (*HPS5*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS6-RELATED (*HPS6*) **negative**
HOLOCARBOXYLASE SYNTHETASE DEFICIENCY (*HLCS*) **negative**
HOMOCYSTINURIA AND MEGALOBLASTIC ANEMIA TYPE CBLG (*MTR*) **negative**
HOMOCYSTINURIA DUE TO DEFICIENCY OF MTHFR (*MTHFR*) **negative**
HOMOCYSTINURIA, CBS-RELATED (*CBS*) **negative**
HOMOCYSTINURIA, Type cblE (*MTRR*) **negative**
HYDROLETHALUS SYNDROME (*HYLS1*) **negative**
HYPER-IGM IMMUNODEFICIENCY (*CD40*) **negative**
HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA (HHH SYNDROME) (*SLC25A15*) **negative**
HYPERPHOSPHATEMIC FAMILIAL TUMORAL CALCINOSIS, GALNT3-RELATED (*GALNT3*) **negative**
HYPOMYELINATING LEUKODYSTROPHY 12 (*VPS11*) **negative**
HYPOPHOSPHATASIA, ALPL-RELATED (*ALPL*) **negative**

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

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

K
KRABBE DISEASE (*GALC*) **negative**

L
LAMELLAR ICHTHYOSIS, TYPE 1 (*TGM1*) **negative**
LARON SYNDROME (*GHR*) **negative**
LEBER CONGENITAL AMAUROSIS 2 (*RPE65*) **negative**
LEBER CONGENITAL AMAUROSIS TYPE AIPL1 (*AIPL1*) **negative**
LEBER CONGENITAL AMAUROSIS TYPE GUCY2D (*GUCY2D*) **negative**
LEBER CONGENITAL AMAUROSIS TYPE TULP1 (*TULP1*) **negative**
LEBER CONGENITAL AMAUROSIS, IQCB1-RELATED/SENIOR-LOKEN SYNDROME 5 (*IQCB1*) **negative**
LEBER CONGENITAL AMAUROSIS, TYPE CEP290 (*CEP290*) **negative**
LEBER CONGENITAL AMAUROSIS, TYPE LCA5 (*LCA5*) **negative**
LEBER CONGENITAL AMAUROSIS, TYPE RDH12 (*RDH12*) **negative**
LEIGH SYNDROME, FRENCH-CANADIAN TYPE (*LRPPRC*) **negative**
LETHAL CONGENITAL CONTRACTURE SYNDROME 1 (*GLE1*) **negative**
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER (*EIF2B5*) **negative**
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B1-RELATED (*EIF2B1*) **negative**
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B2-RELATED (*EIF2B2*) **negative**
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B3-RELATED (*EIF2B3*) **negative**
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B4-RELATED (*EIF2B4*) **negative**
LIG4 SYNDROME (*LIG4*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 8 (*TRIM32*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2A (*CAPN3*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2B (*DYSF*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2C (*SGCG*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2D (*SGCA*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2E (*SGCB*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2F (*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*) **see first page**
MEROSIN-DEFICIENT MUSCULAR DYSTROPHY (*LAMA2*) **negative**
METABOLIC ENCEPHALOPATHY AND ARRHYTHMIAS, TANGO2-RELATED (*TANGO2*) **negative**
METACHROMATIC LEUKODYSTROPHY, ARSA-RELATED (*ARSA*) **negative**
METACHROMATIC LEUKODYSTROPHY, PSAP-RELATED (*PSAP*) **negative**
METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CBLF (*LMBRD1*) **negative**
METHYLMALONIC ACIDEMIA, MCEE-RELATED (*MCEE*) **negative**
METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CBLC (*MMACHC*) **negative**
METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CblD (*MMADHC*) **negative**
METHYLMALONIC ACIDURIA, MMAA-RELATED (*MMAA*) **negative**
METHYLMALONIC ACIDURIA, MMAB-RELATED (*MMAB*) **negative**
METHYLMALONIC ACIDURIA, TYPE MUT(0) (*MUT*) **negative**
MEVALONIC KINASE DEFICIENCY (*MVK*) **negative**
MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II (*PCNT*) **negative**
MICROPHTHALMIA / ANOPHTHALMIA, VSX2-RELATED (*VSX2*) **negative**
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, ACAD9-RELATED (*ACAD9*) **negative**
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFAF5-RELATED (*NDUFAF5*) **negative**
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFS6-RELATED (*NDUFS6*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 1 (*NDUFS4*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 10 (*NDUFAF2*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 17 (*NDUFAF6*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 19 (*FOXRED1*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 3 (*NDUFS7*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 4 (*NDUFV1*) **negative**
MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 2, SCO2-RELATED (*SCO2*) **negative**
MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 6 (*COX15*) **negative**
MITOCHONDRIAL DNA DEPLETION SYNDROME 2 (*TK2*) **negative**
MITOCHONDRIAL DNA DEPLETION SYNDROME 3 (*DGUOK*) **negative**
MITOCHONDRIAL MYOPATHY AND SIDEROBLASTIC ANEMIA (MLASA1) (*PUS1*) **negative**
MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (*HADHB*) **negative**
MOLYBDENUM COFACTOR DEFICIENCY TYPE B (*MOCS2*) **negative**
MOLYBDENUM COFACTOR DEFICIENCY, TYPE A (*MOCS1*) **negative**
MUCOLIPIDOSIS II/III A (*GNPTAB*) **negative**
MUCOLIPIDOSIS III GAMMA (*GNPTG*) **negative**
MUCOLIPIDOSIS, TYPE IV (*MCOLN1*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE I (HURLER SYNDROME) (*IDUA*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III A (SANFILIPPO A) (*SGSH*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III B (SANFILIPPO B) (*NAGLU*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III C (SANFILIPPO C) (*HGSNAT*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III D (SANFILIPPO D) (*GNS*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE IV A (MORQUIO SYNDROME) (*GALNS*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE IV B/GM1 GANGLIOSIDOSIS (*GLB1*) **see first page**
MUCOPOLYSACCHARIDOSIS, TYPE IX (*HYAL1*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE VI (MAROTEAUX-LAMY) (*ARSB*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE VII (*GUSB*) **negative**
MULIBREY NANISM (*TRIM37*) **negative**
MULTIPLE PTERYGIUM SYNDROME, CHRNG-RELATED/ESCOBAR SYNDROME (*CHRNG*) **negative**
MULTIPLE SULFATASE DEFICIENCY (*SUMF1*) **negative**
MUSCLE-EYE-BRAIN DISEASE, POMGNT1-RELATED (*POMGNT1*) **negative**
MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (*RXYLT1*) **negative**
MUSK-RELATED CONGENITAL MYASTHENIC SYNDROME (*MUSK*) **negative**
MYONEUROGASTROINTESTINAL ENCEPHALOPATHY (MNGIE) (*TYMP*) **negative**
MYOTONIA CONGENITA (*CLCN1*) **negative**

N
N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY (*NAGS*) **negative**
NEMALINE MYOPATHY, NEB-RELATED (*NEB*) **negative**
NEPHRONOPHTHISIS 1 (*NPHP1*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, CLN5-RELATED (*CLN5*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, CLN6-RELATED (*CLN6*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, CLN8-RELATED (*CLN8*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, MFSD8-RELATED (*MFSD8*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, PPT1-RELATED (*PPT1*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, TPP1-RELATED (*TPP1*) **negative**
NGLY1-CONGENITAL DISORDER OF GLYCOSYLATION (*NGLY1*) **negative**
NIEMANN-PICK DISEASE, TYPE C1 / D (*NPC1*) **negative**
NIEMANN-PICK DISEASE, TYPE C2 (*NPC2*) **negative**
NIEMANN-PICK DISEASE, TYPES A / B (*SMPD1*) **negative**
NIJMEGEN BREAKAGE SYNDROME (*NBN*) **negative**
NON-SYNDROMIC HEARING LOSS, GJB2-RELATED (*GJB2*) **negative**
NON-SYNDROMIC HEARING LOSS, MYO15A-RELATED (*MYO15A*) **negative**
NONSYNDROMIC HEARING LOSS, OTOA-RELATED (*OTOA*) **negative**
NONSYNDROMIC HEARING LOSS, OTOF-RELATED (*OTOF*) **negative**
NONSYNDROMIC HEARING LOSS, PJVK-RELATED (*PJVK*) **negative**
NONSYNDROMIC HEARING LOSS, SYNE4-RELATED (*SYNE4*) **negative**
NONSYNDROMIC HEARING LOSS, TMC1-RELATED (*TMC1*) **negative**
NONSYNDROMIC HEARING LOSS, TMPRSS3-RELATED (*TMPRSS3*) **negative**
NONSYNDROMIC INTELLECTUAL DISABILITY (*CC2D1A*) **negative**
NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (*SAMD9*) **negative**

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

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

P
POLG-RELATED DISORDERS (*POLG*) **negative**
POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE (*PKHD1*) **negative**
PONTOCEREBELLAR HYPOPLASIA, EXOSC3-RELATED (*EXOSC3*) **negative**
PONTOCEREBELLAR HYPOPLASIA, RARS2-RELATED (*RARS2*) **negative**
PONTOCEREBELLAR HYPOPLASIA, TSEN2-RELATED (*TSEN2*) **negative**
PONTOCEREBELLAR HYPOPLASIA, TSEN54-RELATED (*TSEN54*) **negative**
PONTOCEREBELLAR HYPOPLASIA, TYPE 1A (*VRK1*) **negative**
PONTOCEREBELLAR HYPOPLASIA, TYPE 2D (*SEPSECS*) **negative**
PONTOCEREBELLAR HYPOPLASIA, VPS53-RELATED (*VPS53*) **negative**
PRIMARY CILIARY DYSKINESIA, CCDC103-RELATED (*CCDC103*) **negative**
PRIMARY CILIARY DYSKINESIA, CCDC39-RELATED (*CCDC39*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAH11-RELATED (*DNAH11*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAH5-RELATED (*DNAH5*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAI1-RELATED (*DNAI1*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAI2-RELATED (*DNAI2*) **negative**
PRIMARY CONGENITAL GLAUCOMA/PETERS ANOMALY (*CYP1B1*) **negative**
PRIMARY HYPEROXALURIA, TYPE 1 (*AGXT*) **negative**
PRIMARY HYPEROXALURIA, TYPE 2 (*GRHPR*) **negative**
PRIMARY HYPEROXALURIA, TYPE 3 (*HOGA1*) **negative**
PRIMARY MICROCEPHALY 1, AUTOSOMAL RECESSIVE (*MCPH1*) **negative**
PROGRESSIVE EARLY-ONSET ENCEPHALOPATHY WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM (*TBCD*) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, ABCB4-RELATED (*ABCB4*) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 1 (PFIC1) (*ATP8B1*) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 2 (*ABCB11*) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 4 (PFIC4) (*TJP2*) **negative**
PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA (*CCN6*) **negative**
PROLIDASE DEFICIENCY (*PEPD*) **negative**
PROPIONIC ACIDEMIA, PCCA-RELATED (*PCCA*) **negative**
PROPIONIC ACIDEMIA, PCCB-RELATED (*PCCB*) **negative**
PSEUDOXANTHOMA ELASTICUM (*ABCC6*) **negative**
PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (PCD) DEFICIENCY (*PCBD1*) **negative**
PYCNODYSOSTOSIS (*CTSK*) **negative**
PYRIDOXAL 5'-PHOSPHATE-DEPENDENT EPILEPSY (*PNPO*) **negative**
PYRIDOXINE-DEPENDENT EPILEPSY (*ALDH7A1*) **negative**
PYRUVATE CARBOXYLASE DEFICIENCY (*PC*) **negative**
PYRUVATE DEHYDROGENASE DEFICIENCY, PDHB-RELATED (*PDHB*) **negative**

R
REFSUM DISEASE, PHYH-RELATED (*PHYH*) **negative**
RENAL TUBULAR ACIDOSIS AND DEAFNESS, ATP6V1B1-RELATED (*ATP6V1B1*) **negative**
RENAL TUBULAR ACIDOSIS, PROXIMAL, WITH OCULAR ABNORMALITIES AND MENTAL RETARDATION (*SLC4A4*) **negative**
RETINITIS PIGMENTOSA 25 (*EYS*) **negative**
RETINITIS PIGMENTOSA 26 (*CERKL*) **negative**
RETINITIS PIGMENTOSA 28 (*FAM161A*) **negative**
RETINITIS PIGMENTOSA 36 (*PRCD*) **negative**
RETINITIS PIGMENTOSA 59 (*DHDDS*) **negative**
RETINITIS PIGMENTOSA 62 (*MAK*) **negative**
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1 (*PEX7*) **negative**
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2 (*GNPAT*) **negative**
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3 (*AGPS*) **negative**
RLBP1-RELATED RETINOPATHY (*RLBP1*) **negative**
ROBERTS SYNDROME (*ESCO2*) **negative**
RYR1-RELATED CONDITIONS (*RYR1*) **negative**

S
SALLA DISEASE (*SLC17A5*) **negative**
SANDHOFF DISEASE (*HEXB*) **negative**
SCHIMKE IMMUNOOSSEOUS DYSPLASIA (*SMARCAL1*) **negative**
SCHINDLER DISEASE (*NAGA*) **negative**
SEGAWA SYNDROME, TH-RELATED (*TH*) **negative**
SENIOR-LOKEN SYNDROME 4/NEPHRONOPHTHISIS 4 (*NPHP4*) **negative**
SEPIAPTERIN REDUCTASE DEFICIENCY (*SPR*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3D-RELATED (*CD3D*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3E-RELATED (*CD3E*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), FOXN1-RELATED (*FOXN1*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), IKBKB-RELATED (*IKBKB*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), IL7R-RELATED (*IL7R*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), JAK3-RELATED (*JAK3*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), PTPRC-RELATED (*PTPRC*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), RAG1-RELATED (*RAG1*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY, ADA-Related (*ADA*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY, TYPE ATHABASKAN (*DCLRE1C*) **negative**
SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY (*DYNC2H1*) **negative**
SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED (*SBDS*) **negative**
SIALIDOSIS (*NEU1*) **negative**
SJÖGREN-LARSSON SYNDROME (*ALDH3A2*) **negative**
SMITH-LEMLI-OPITZ SYNDROME (*DHCR7*) **negative**
SPASTIC PARAPLEGIA, TYPE 15 (*ZFYVE26*) **negative**
SPASTIC TETRAPLEGIA, THIN CORPUS CALLOSUM, AND PROGRESSIVE MICROCEPHALY (SPATCCM) (*SLC1A4*) **negative**
SPG11-RELATED CONDITIONS (*SPG11*) **negative**
SPINAL MUSCULAR ATROPHY (*SMN1*) **negative SMN1: Two copies; g.27134T>G: absent; the absence of the g.27134T>G variant decreases the chance to be a silent (2+0) carrier.**
SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS TYPE 1 (*IGHMBP2*) **negative**
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 10 (*ANO10*) **negative**
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12 (*WWOX*) **negative**
SPONDYLOCOSTAL DYSOSTOSIS 1 (*DLL3*) **negative**
SPONDYLOTHORACIC DYSOSTOSIS, MESP2-Related (*MESP2*) **negative**
STEEL SYNDROME (*COL27A1*) **negative**
STEROID-RESISTANT NEPHROTIC SYNDROME (*NPHS2*) **negative**
STUVE-WIEDEMANN SYNDROME (*LIFR*) **negative**
SURF1-RELATED CONDITIONS (*SURF1*) **negative**
SURFACTANT DYSFUNCTION, ABCA3-RELATED (*ABCA3*) **negative**

T
TAY-SACHS DISEASE (*HEXA*) **negative**
TBCE-RELATED CONDITIONS (*TBCE*) **negative**
THIAMINE-RESPONSIVE MEGALOBLASTIC ANEMIA SYNDROME (*SLC19A2*) **negative**
THYROID DYSHORMONOGENESIS 1 (*SLC5A5*) **negative**
THYROID DYSHORMONOGENESIS 2A (*TPO*) **negative**
THYROID DYSHORMONOGENESIS 3 (*TG*) **negative**
THYROID DYSHORMONOGENESIS 6 (*DUOX2*) **negative**
TRANSCOBALAMIN II DEFICIENCY (*TCN2*) **negative**
TRICHOHEPATOENTERIC SYNDROME, SKIC2-RELATED (*SKIC2*) **negative**
TRICHOHEPATOENTERIC SYNDROME, TTC37-RELATED (*TTC37*) **negative**
TRICHOTHIODYSTROPHY 1/XERODERMA PIGMENTOSUM, GROUP D (*ERCC2*) **negative**
TRIMETHYLAMINURIA (*FMO3*) **negative**
TRIPLE A SYNDROME (*AAAS*) **negative**
TSHR-RELATED CONDITIONS (*TSHR*) **negative**
TYROSINEMIA TYPE III (*HPD*) **negative**
TYROSINEMIA, TYPE 1 (*FAH*) **negative**
TYROSINEMIA, TYPE 2 (*TAT*) **negative**

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

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

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

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

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

Patient Information
Patient Name:

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

Test Information
Ordering Physician: [redacted]

Clinic Information:

Report Date:

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

Patient Information
Patient Name:

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

Test Information
Ordering Physician: [redacted]

Clinic Information:

Report Date:

horizon™ natera carrier screen

Testing Methodology, Limitations, and Comments:

Next-generation sequencing (NGS)

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

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

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

SPECIAL NOTES

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

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

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

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

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

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

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

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

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

Friedreich Ataxia (FXN)

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

Friedreich Ataxia Repeat Categories

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

Patient Information
Patient Name:

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

Test Information
Ordering Physician: [redacted]

Clinic Information:

Report Date:

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.