



## Donor 7624

### Genetic Testing Summary

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

Last Updated: 09/18/24

Donor Reported Ancestry: English, Irish

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.	<p>Carrier: Congenital Myasthenic Syndrome, CHRNE-Related</p> <p>Carrier: Oculocutaneous Albinism, OCA2-Related</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 7624

Date Of Birth:

Gender: Male

 Ethnicity: Northern European  
Caucasian

Patient ID:

Medical Record #: N/A

Collection Kit:

Accession ID: N/A

Case File ID:

**Test Information**

Ordering Physician:

Clinic Information: Fairfax Cryobank

Phone:

Report Date: 06/26/2024

Sample Collected: 06/12/2024

Sample Received: 06/13/2024

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 Myasthenic Syndrome, CHRNE-Related**

Positive for the pathogenic variant c.295C>T (p.R99\*) in the CHRNE gene. Although most variants in this gene are associated with an autosomal recessive form of CONGENITAL MYASTHENIC SYNDROME, CHRNE-RELATED, some rare CHRNE variants may cause an autosomal dominant form of the condition. To our knowledge, there is insufficient evidence that this variant causes an autosomal dominant form of this condition. If this individual's partner is a carrier for this CONGENITAL MYASTHENIC SYNDROME, CHRNE-RELATED, their chance to have a child with this condition is likely 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

**CARRIER for Oculocutaneous Albinism, OCA2-Related**

Positive for the pathogenic variant c.1327G>A (p.V443I) in the OCA2 gene. If this individual's partner is a carrier for OCULOCUTANEOUS ALBINISM, OCA2-RELATED, their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

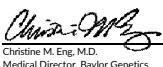
**Negative for 547 out of 549 diseases**

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

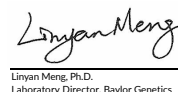
Carrier screening is not diagnostic and may not detect all possible pathogenic variants in a given gene.

**RECOMMENDATIONS**

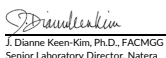
Individuals who would like to review their Horizon report with a Natera Laboratory Genetic Counselor may schedule a telephone genetic information session by calling 650-249-9090 or visiting [naterasession.com](https://www.naterasession.com). Clinicians with questions may contact Natera at 650-249-9090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



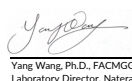
Christine M. Eng, M.D.  
Medical Director, Baylor Genetics



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



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



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Laboratory Director, Natera

**Patient Information**

Patient Name: Donor 7624

**Test Information**

Ordering Physician: [REDACTED]



Clinic Information: Fairfax Cryobank

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: 06/26/2024

**CONGENITAL MYASTHENIC SYNDROME, CHRNE-RELATED****Understanding Your Horizon Carrier Screen Results****What is Congenital Myasthenic Syndrome, CHRNE-Related?**

Congenital Myasthenic Syndrome, CHRNE-Related is an inherited disorder that affects the muscles. Symptoms of muscle weakness (myasthenia) can begin after birth but may begin later in life. Affected infants and children often have feeding and swallowing problems, developmental delay, and at times may have breathing problems. Muscle weakness can worsen with exercise. Speech problems may occur due to facial muscle weakness. The weakness remains stable and does not worsen with age. The degree of muscle weakness varies among individuals affected with Congenital Myasthenic Syndrome, CHRNE-Related. Clinical trials involving potential new treatments for this condition may be available (see [www.clinicaltrials.gov](http://www.clinicaltrials.gov)).

**What causes Congenital Myasthenic Syndrome, CHRNE-Related?**

The majority of cases of Congenital Myasthenic Syndrome, CHRNE-Related are caused by a change, or mutation, in both copies of the CHRNE gene pair. These mutations cause the genes to not work properly or not work at all. The function of the CHRNE genes is to help relay signals from the nerve cells to the muscle cells; these signals trigger muscle movement. When both copies of the CHRNE gene do not work correctly, the signals from the nerves to the muscles are disrupted, causing problems with movement of skeletal muscles, muscle weakness, and delayed development of motor skills. Most cases of Congenital Myasthenic Syndrome, CHRNE-Related 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 CHRNE gene to have a child with this type of Congenital Myasthenic Syndrome. People who are carriers for Congenital Myasthenic Syndrome, CHRNE-Related are usually healthy and do not have symptoms of the disorder. 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 Congenital Myasthenic Syndrome, CHRNE-Related, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their CHRNE gene mutations to the child, who will then have the disorder. Individuals found to carry more than one mutation for Congenital Myasthenic Syndrome, CHRNE-Related should discuss their risk for having an affected child, and any potential effects to their own health, with their health care provider. Some cases of late-onset Congenital Myasthenic Syndrome, CHRNE-Related are inherited in an autosomal dominant manner. This means that a person who has a mutation in just one copy of the CHRNE gene will have symptoms of the condition. A person with the autosomal dominant form of Congenital Myasthenic Syndrome, CHRNE-Related would have a 1 in 2, or 50%, chance with each pregnancy to pass the mutation to the child, who would then have the autosomal dominant form of this condition. It is sometimes, but not always, possible to determine whether a specific mutation in the CHRNE gene will cause the autosomal recessive or autosomal dominant form of this condition. There are a number of other forms of Congenital Myasthenic Syndrome, each caused by mutations in different genes. A person who is a carrier for Congenital Myasthenic Syndrome, CHRNE-Related is not likely to be at increased risk for having children with these other forms.

**What can I do next?**

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website ([www.nsgc.org](http://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 Congenital Myasthenic Syndrome, CHRNE-Related ordered by a health care professional. If your partner is not found to be a carrier for Congenital Myasthenic Syndrome, CHRNE-Related, your risk of having a child with this disorder is greatly reduced. Couples at risk of having a baby with Congenital Myasthenic Syndrome, CHRNE-Related can opt to have prenatal diagnosis done through chorionic villus sampling (CVS) or amniocentesis during pregnancy or can choose to have the baby tested after birth for this condition. If you are not yet pregnant, your partner can have carrier screening for Congenital Myasthenic Syndrome, CHRNE-Related ordered by a health care professional. If your partner is found to be a carrier for this 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 Congenital Myasthenic Syndrome, CHRNE-Related
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test the embryos for Congenital Myasthenic Syndrome, CHRNE-Related
- Adoption or use of a sperm or egg donor who is not a carrier for Congenital Myasthenic Syndrome, CHRNE-Related

**What resources are available?**

- Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/congenital-myasthenic-syndrome>
- 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]

Case File ID: [REDACTED]

Report Date: [REDACTED]

**OCULOCUTANEOUS ALBINISM, OCA2-RELATED****Understanding Your Horizon Carrier Screen Results****What is Oculocutaneous Albinism, OCA2-Related?**

Oculocutaneous Albinism, OCA2-Related, is an inherited disorder that affects the pigmentation (coloring) of the eyes, skin, and hair. People with Oculocutaneous Albinism, OCA2-Related, are born with less melanin, the substance that creates body coloring. This leads to lighter than average color of the hair, skin, and eyes, especially at birth and in infancy. Some people with this condition produce more pigment over time, leading to skin, hair, and eye color that is closer to that typical for their family. Some affected people have vision problems that can include light sensitivity (photophobia), involuntary eye movements (nystagmus), and blurry vision (decreased acuity).

Currently there is no cure for this condition and treatment is based on symptoms. Treatments may include avoiding sun exposure and use of eyeglasses, sunglasses, and other vision aids. Clinical trials involving potential new treatments for this condition may be available (see [www.clinicaltrials.gov](http://www.clinicaltrials.gov)).

**What causes Oculocutaneous Albinism, OCA2-Related?**

Oculocutaneous Albinism, OCA2-Related, is caused by a change, or mutation, in both copies of the OCA2 gene pair. These mutations cause the gene to not work properly or not work at all. The job of the OCA2 gene is to help make melanin, which determines the coloring of our eyes, skin, and hair. When both copies of this gene are not working correctly, it leads to the symptoms described above.

Oculocutaneous Albinism, OCA2-Related, is inherited in an autosomal recessive manner. This means that, in most cases, both parents must be carriers of a mutation in one copy of the OCA2 gene to have a child with this condition. People who are carriers of Oculocutaneous Albinism, OCA2-Related, are usually healthy and do not have the condition themselves. Usually a child inherits two copies of each gene, one copy from their mother and one copy from their father. If the mother and father are both carriers of Oculocutaneous Albinism, OCA2-Related, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their OCA2 gene mutations to a child, who will then have this condition.

Individuals found to carry more than one mutation for Oculocutaneous Albinism, OCA2-Related, should discuss their risk for having an affected child with their healthcare provider.

**What can I do next?**

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website ([www.nsgc.org](http://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 OCA2 mutations ordered by a healthcare professional. If your partner is not found to be a carrier of an OCA2 mutation, the chance that you would have a child with Oculocutaneous Albinism, OCA2-Related, is very low and no further testing would be recommended. If your partner also carries an OCA2 mutation, and there is a 1 in 4, or 25%, chance of having an affected child, you can choose to test the pregnancy with chorionic villus sampling (CVS) or amniocentesis or you can have the baby tested after birth for this condition.

If you are not yet pregnant, your partner can have carrier screening for OCA2 mutations ordered by a healthcare professional. If your partner is also a carrier of Oculocutaneous Albinism, OCA2-Related, and your future children each have a 1 in 4, or 25%, chance of having Oculocutaneous Albinism, OCA2-Related, you have several reproductive options to consider:

- Natural pregnancy with or without prenatal diagnosis of the fetus or testing the baby after birth for Oculocutaneous Albinism, OCA2-Related,
- Preimplantation genetic testing (PGT) with in vitro fertilization (IVF) to test embryos for Oculocutaneous Albinism, OCA2-Related, or
- Adoption or use of a sperm or egg donor who is not a carrier for Oculocutaneous Albinism, OCA2-Related.

**What resources are available?**

- MedlinePlus: [medlineplus.gov/genetics/condition/oculocutaneous-albinism/](http://medlineplus.gov/genetics/condition/oculocutaneous-albinism/)
- National Organization for Rare Disorders [rarediseases.org/rare-diseases/oculocutaneous-albinism/](http://rarediseases.org/rare-diseases/oculocutaneous-albinism/)
- National Organization for Albinism and Hypopigmentation [www.albinism.org](http://www.albinism.org)
- Prenatal diagnosis done through CVS [www.marchofdimes.org/chorionic-villus-sampling.aspx](http://www.marchofdimes.org/chorionic-villus-sampling.aspx)
- Prenatal diagnosis done through amniocentesis [www.marchofdimes.org/amniocentesis.aspx](http://www.marchofdimes.org/amniocentesis.aspx)
- Preimplantation genetic diagnosis (PGD) with IVF [www.natera.com/spectrum](http://www.natera.com/spectrum)

**Patient Information**

Patient Name: [REDACTED]

**Test Information**

Ordering Physician: [REDACTED]



Date Of Birth: [REDACTED]

Clinic Information:

Case File ID: [REDACTED]

Report Date:

**VARIANT DETAILS****CHRNE, c.295C>T (p.R99\*), heterozygous, pathogenic**

- The c.295C>T (p.R99\*) variant in the CHRNE gene has been observed at a frequency of 0.0004% 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 congenital myasthenic syndrome (PMID: 29702980).
- This premature termination variant is predicted to cause nonsense-mediated decay (NMD) in a gene where loss-of-function is a known mechanism of disease.
- This variant has been reported in ClinVar [ID: 1399349].

**OCA2, c.1327G>A (p.V443I), heterozygous, pathogenic**

- The c.1327G>A (p.V443I) variant in the OCA2 gene has been observed at a frequency of 0.3056% 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 oculocutaneous albinism, type II (PMID: 8302318, 15712365, 18326704, 18463683).
- This variant has been reported in ClinVar [ID: 955].

**Patient Information**

Patient Name:

**Test Information**

Ordering Physician:



Clinic Information:

Date Of Birth:

Case File ID:

Report Date:

**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-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**  
ATRAFERRINEMIA (*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**  
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 PALMOPANTAR 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**  
CHOREOACANTHOCTOSIS (*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 (*TSMF*) **negative**  
COMBINED PITUITARY HORMONE DEFICIENCY 1 (*POU1F1*) **negative**  
COMBINED PITUITARY HORMONE DEFICIENCY-2 (*PROP1*) **negative**  
CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (*CYP11B1*) **negative**  
CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (*CYP17A1*) **negative**  
CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (*CYP21A2*) **negative**  
CONGENITAL ADRENAL INSUFFICIENCY, CYP11A1-RELATED (*CYP11A1*) **negative**  
CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (*MPL*) **negative**  
CONGENITAL CHRONIC DIARRHEA (*DGAT1*) **negative**  
CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, ALG1-RELATED (*ALG1*) **negative**  
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-Related (*PMM2*) **negative**  
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (*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*) **see first page**  
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*) **negative**  
CYSTINOSIS (*CTNS*) **negative**  
CYTOCHROME C OXIDASE DEFICIENCY, PET100-RELATED (*PET100*) **negative**  
CYTOCHROME P450 OXIOREDUCTASE DEFICIENCY (*POR*) **negative**
- D**  
D-BIFUNCTIONAL PROTEIN DEFICIENCY (*HSD17B4*) **negative**

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**D**

DEAFNESS, AUTOSOMAL RECESSIVE 77 (*LOXHD1*) **negative**  
 DIHYDROPTERIDINE REDUCTASE (DHPR) DEFICIENCY (*QDPR*) **negative**  
 DONNAI-BARROW SYNDROME (*LRP2*) **negative**  
 DUBIN-JOHNSON SYNDROME (*ABCC2*) **negative**  
 DYSKERATOSIS CONGENITA SPECTRUM DISORDERS (*TERT*) **negative**  
 DYSKERATOSIS CONGENITA, RTTEL1-RELATED (*RTTEL1*) **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 (*HLC5*) **negative**  
 HOMOCYSTEINURIA AND MEGALOBlastic ANEMIA TYPE CBLG (*MTR*) **negative**  
 HOMOCYSTEINURIA DUE TO DEFICIENCY OF MTHFR (*MTHFR*) **negative**  
 HOMOCYSTEINURIA, CBS-RELATED (*CBS*) **negative**  
 HOMOCYSTEINURIA, 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 (*JNV5*) **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, AH11-RELATED (*AH11*) **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/LARYNGOOXYCHOCUTANEOUS SYNDROME, LAMA3-RELATED (*LAMA3*) **negative**

**K**

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

**L**

LAMELLAR ICHTHYOSIS, TYPE 1 (*TGM1*) **negative**

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

**M**  
 MALONYL-CoA DECARBOXYLASE DEFICIENCY (*MLYCD*) **negative**  
 MAPLE SYRUP URINE DISEASE, TYPE 1A (*BCKDHA*) **negative**  
 MAPLE SYRUP URINE DISEASE, TYPE 1B (*BCKDHB*) **negative**  
 MAPLE SYRUP URINE DISEASE, TYPE 2 (*DBT*) **negative**  
 MCKUSICK-KAUFMAN SYNDROME (*MKKS*) **negative**  
 MECKEL SYNDROME 7/NEPHRONOPHTHISIS 3 (*NPHP3*) **negative**  
 MECKEL-GRUBER SYNDROME, TYPE 1 (*MKS1*) **negative**  
 MECR-RELATED NEUROLOGIC DISORDER (*MECR*) **negative**  
 MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (*ACADM*) **negative**  
 MEDNIK SYNDROME (*AP1S1*) **negative**  
 MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS (*MLC1*) **negative**  
 MEROSIN-DEFICIENT MUSCULAR DYSTROPHY (*LAMA2*) **negative**  
 METABOLIC ENCEPHALOPATHY AND ARRHYTHMIAS, TANGO2-RELATED (*TANGO2*) **negative**  
 METACHROMATIC LEUKODYSTROPHY, ARSA-RELATED (*ARSA*) **negative**  
 METACHROMATIC LEUKODYSTROPHY, PSAP-RELATED (*PSAP*) **negative**  
 METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CBLF (*LMBRD1*) **negative**  
 METHYLMALONIC ACIDEMIA, MCEE-RELATED (*MCEE*) **negative**  
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CBLC (*MMACHC*) **negative**  
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CblD (*MMADHC*) **negative**  
 METHYLMALONIC ACIDURIA, MMAA-RELATED (*MMAA*) **negative**  
 METHYLMALONIC ACIDURIA, MMAB-RELATED (*MMAB*) **negative**  
 METHYLMALONIC ACIDURIA, TYPE MUT (0) (*MUT*) **negative**  
 MEVALONIC KINASE DEFICIENCY (*MVK*) **negative**  
 MICROCEPHALIC OSTEOPLASTIC PRIMORDIAL DWARFISM TYPE II (*PCNT*) **negative**  
 MICROPTHALMIA / ANOPHTHALMIA, VSX2-RELATED (*VSX2*) **negative**  
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, ACAD9-RELATED (*ACAD9*) **negative**  
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFAF5-RELATED (*NDUFAF5*) **negative**  
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFS6-RELATED (*NDUFS6*) **negative**  
 MITOCHONDRIAL COMPLEX 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 (*MOC52*) **negative**  
 MOLYBDENUM COFACTOR DEFICIENCY, TYPE A (*MOC51*) **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 VI (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**  
 NONSYNDROMIC HEARING LOSS, OTOF-RELATED (*OTOF*) **negative**  
 NONSYNDROMIC HEARING LOSS, PJK-RELATED (*PJK*) **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 IV (*SLC45A2*) **negative**  
 OCULOCUTANEOUS ALBINISM TYPE, III (*TYRP1*) **negative**  
 OCULOCUTANEOUS ALBINISM, OCA2-RELATED (*OCA2*) **see first page**  
 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**  
 OSTEOPECTOSIS, INFANTILE MALIGNANT, TCIRG1-RELATED (*TCIRG1*) **negative**  
 OSTEOPECTOSIS, OSTM1-RELATED (*OSTM1*) **negative**

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



**Patient Information**

Patient Name: [REDACTED]

**Test Information**

Ordering Physician: [REDACTED]



Clinic Information: [REDACTED]

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: [REDACTED]

**P**

POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE (*PKHD1*) **negative**  
 PONTOCEREBELLAR HYPOPLASIA, EXOSC3-RELATED (*EXOSC3*) **negative**  
 PONTOCEREBELLAR HYPOPLASIA, RARS2-RELATED (*RARS2*) **negative**  
 PONTOCEREBELLAR HYPOPLASIA, TSEN2-RELATED (*TSEN2*) **negative**  
 PONTOCEREBELLAR HYPOPLASIA, TSEN54-RELATED (*TSEN54*) **negative**  
 PONTOCEREBELLAR HYPOPLASIA, TYPE 1A (*VRK1*) **negative**  
 PONTOCEREBELLAR HYPOPLASIA, TYPE 2D (*SEPSEC5*) **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**  
 PYCNODYSTOSIS (*CTS5*) **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 IMMUNOSSESQUOUS 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), FOXP1-RELATED (*FOXP1*) **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**  
 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 (*AAAS*) **negative**  
 TSHR-RELATED CONDITIONS (*TSHR*) **negative**  
 TYROSINEMIA TYPE III (*HPD*) **negative**  
 TYROSINEMIA, TYPE 1 (*FAH*) **negative**  
 TYROSINEMIA, TYPE 2 (*TAT*) **negative**

**U**

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

**V**

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

**W**

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

**X**

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

**Z**

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

**Patient Information**

Patient Name:

**Test Information**

Ordering Physician:



Date Of Birth:



Clinic Information:

Case File ID:



Report Date:

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

**Patient Information**

Patient Name: [REDACTED]

**Test Information**

Ordering Physician: [REDACTED]



Clinic Information:

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

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]

Clinic Information:

Case File ID: [REDACTED]

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.

PATIENT INFORMATION

**DONOR, 7624**

REPORT STATUS **Final**

Nichols Institute, Chantilly

DOB: [REDACTED] Age: [REDACTED]  
SEX: M

ORDERING PHYSICIAN

CLIENT INFORMATION

ID: 7624-[REDACTED]  
PHONE: [REDACTED]

SPECIMEN INFORMATION

SPECIMEN: [REDACTED]  
REQUISITION: [REDACTED]  
LAB REF NO: [REDACTED]

COLLECTED: 06/12/2024 15:00  
RECEIVED: 06/13/2024 14:29  
REPORTED: 06/21/2024 12:53

Test Name	In Range	Out of Range	Reference Range	Lab
Hemoglobinopathy Evaluation				AMD
Red Blood Cell Count	5.64		4.20-5.80 Mill/uL	
HEMOGLOBIN	16.4		13.2-17.1 g/dL	
Hematocrit				
<b>Hematocrit</b>		<b>51.8 H</b>	38.5-50.0 %	
MCV	91.8		80.0-100.0 fL	
MCH	29.1		27.0-33.0 pg	
RDW	12.0		11.0-15.0 %	
Hemoglobin A	97.7		>96.0 %	
Hemoglobin F	0.0		<2.0 %	
Hemoglobin A2 (Quant)	2.3		2.0-3.2 %	
Interpretation				

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.

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

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[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

PATIENT INFORMATION

DONOR, 7624

REPORT STATUS **Final**

Nichols Institute, Chantilly

ORDERING PHYSICIAN

DOB: [REDACTED] Age: [REDACTED]

SEX: M

ID: 7624 [REDACTED]

COLLECTED: 06/12/2024 15:00

REPORTED: 06/21/2024 12:53

Test Name	In Range	Out of Range	Reference Range	Lab
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]
[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]	[REDACTED]

Chromosome Analysis, Blood AMD  
 Chromosome Analysis, Blood  
 Chromosome Analysis, Blood

Order ID: [REDACTED]

Specimen Type: Blood

Clinical Indication: Gamete donor

RESULT:  
NORMAL MALE KARYOTYPE

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

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

NOMENCLATURE:  
46,XY

ASSAY INFORMATION:  
 Method: G-Band (Digital Analysis:  
 MetaSystems/Ikaros)  
 Cells Counted: 20  
 Band Level: 550  
 Cells Analyzed: 6  
 Cells Karyotyped: 6

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.

Steven A. Schonberg, Ph.D., FACMG, Technical Director, Cytogenetics

PATIENT INFORMATION

**DONOR, 7624**

REPORT STATUS **Final**

Nichols Institute, Chantilly

ORDERING PHYSICIAN

DOB: [REDACTED]

Age: [REDACTED]

SEX: M

ID: 7624 [REDACTED]

COLLECTED: 06/12/2024 15:00

REPORTED: 06/21/2024 12:53

Test Name	In Range	Out of Range	Reference Range	Lab
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Chromosome Analysis, Blood (Continued)				
Chromosome Analysis, Blood (Continued)				
and Genomics, 703-802-7156				

Electronic Signature: 6/21/2024 12:10 PM

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

**Performing Laboratory Information:**

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