Reproductive Technologies, Inc.
THE SPERM BANK OF CALIFORNIA
2115 MILVIA STREET, BERKELEY 94704 PHONE 510.841.1858 www.thespermbankofca.org A 501(c)(3) CORPORATION

# Acknowledgement of Positive Carrier Screening Results: Donor 5419

I, the undersigned recipient, understand that this donor has tested **POSITIVE** as a carrier for the following condition(s):

- **Glycine Encephalopathy- GLDC related**

I intend to use sperm samples from this donor for insemination or other assisted conception procedure(s).

I acknowledge that The Sperm Bank of California (TSBC) has made the donor's genetic testing results available to me and my medical providers, and that I have reviewed these results. I understand that TSBC **strongly recommends** that I review these genetic testing results with a Genetic Counselor and my medical providers. I understand that TSBC can refer me to genetic counseling services if desired.

**I understand that recipient testing is strongly recommended when a donor has positive carrier screening results and that such testing can reduce but not eliminate risks.**

I acknowledge that I personally assume all risks associated with use of semen samples provided by a donor who has tested **POSITIVE as a carrier for Glycine Encephalopathy- GLDC related.**

On behalf of myself and my spouse, heirs, representatives, I hereby release and forever hold harmless TSBC and its current and former officers, directors, employees, attorneys, insurers, consultants, agents, and representatives (collectively "Releases") from any liability or responsibility whatsoever for any and all outcomes, and hereby release and forever discharge Releases from any and all actions, causes of action, demands, damages, losses, liabilities, suits, expenses, including attorneys' fees and costs, of whatever character, in law or in equity, whether currently known, suspected, unknown or unsuspected, matured or unmatured, arising out of my use of sperm donated by a donor who has tested **POSITIVE as a carrier for Glycine Encephalopathy-GLDC related. This release involves the waiver of all rights and benefits that I may have under California Civil Code section 1542, which states: "A general release does not extend to claims that the creditor or releasing party does not know or suspect to exist in his or her favor at the time of executing the release and that, if known by him or her, would have materially affected his or her settlement with the debtor or released party."**

**Please select one of the following:**

- ☐ I have been tested for the above named condition(s) and/or I plan to be tested prior to using the samples.
- ☐ I understand that TSBC **strongly recommends** that I discuss these results with a Genetic Counselor and my medical providers and consider testing for the above named condition(s). At this time I have **declined** testing and/or **do not anticipate being tested**.

I understand that if I transfer my vials (or embryos if applicable) to any other person, including my spouse, that TSBC requires that person (1) register with TSBC and (2) complete an **Acknowledgement of Positive Carrier Screening Results**.

I understand that any and all questions as to the legal interpretation, validity or any other aspect of this agreement shall be determined by the laws of the State of California, regardless of the location or residence of any of the parties.

| Recipient's signature | Recipient's printed name | Date |
|---|---|---|

Reproductive Technologies, Inc.
THE SPERM BANK OF CALIFORNIA
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## GENETIC TESTING: POSITIVE CARRIER STATUS

This donor tested **POSITIVE** as a carrier for one or more autosomal recessive conditions as described on the prior page and in the attached genetic testing results.

### *What does it mean to be a carrier?*

All people carry genetic mutations in their DNA. Genetic testing can help to identify some, but not all, of these mutations. While this donor carries a mutation for one or more recessively inherited condition(s), **offspring from this donor are not expected to be at risk of developing these condition(s) unless the recipient (or egg provider if different from the recipient) also carries a genetic mutation for the same condition(s).** For this reason, we strongly encourage you to discuss carrier screening for yourself (or your egg provider) with your physician and a genetic counselor. Genetic testing can reduce but not eliminate risks.

### *What are my next steps?*

1. **Download the genetic test results and review with your medical providers**
   We **strongly recommend** that you discuss this donor's genetic test results with your physician **PRIOR TO SCHEDULING A SHIPMENT OR PICK-UP**, to confirm the donor is suitable for your use. Vials retrieved from the building cannot be exchanged or refunded. The donor's genetic test results are available for free download on the donor's page at https://www.thespermbankofca.org/donor-catalog.

2. **We recommend scheduling a genetic counseling session.**
   A genetic counselor can explain the results in detail including the inheritance pattern, potential risks to your children, and the available testing options that you may want to consider for yourself (or your egg provider). Phone or in person consultations are available for a fee with TSBC's Genetic Counselors at San Francisco Genetic Counseling (https://www.sfgenetics.org/) or you can locate a genetic counselor at www.findageneticcounselor.com.

3. **Complete and return the Acknowledgement of Positive Carrier Screening Results**
   TSBC requires that all recipients selecting this donor complete this acknowledgement form **PRIOR TO SCHEDULING A SHIPMENT OR PICK-UP**. Completing this form documents that you have been informed about this donor's genetic test results and that you are aware of TSBC's recommendation to discuss the genetic test results with your medical providers as noted above.

**DATE: 08/18/2023**

Reproductive Technologies, Inc.

# THE SPERM BANK OF CALIFORNIA

## EXPANDED CARRIER SCREENING RESULTS DONOR 5419

Expanded carrier screening for 400 autosomal recessive conditions was completed by Horizon Natera Screen and reported to TSBC August 2023. The results were **POSITIVE** for **Glycine Encephalopathy- GLDC related**. Donor 5419 is a carrier for these conditions.

**It is strongly recommended that recipients who use this donor's sperm undergo carrier screening for these specific conditions.**

Testing was negative for the remainder of genes screened.

| Disease | Result | Residual risk to be a carrier (based on European ancestry) |
|---|---|---|
| **Glycine encephalopathy (GLDC)** | POSITIVE | n/a |
| **Cystic Fibrosis** | Negative | 1 in 481 |
| **Spinal Muscular Atrophy** | Negative: 2 copies exon 7 c.*3+80T>G variant not detected | 1 in 769 |
| **HBB Hemoglobinopathies** | Negative | 1 in 7441 |
| **Alpha Thalassemia** | Negative | 1 in 4991 |

Genetic screening tests can significantly reduce, but never completely eliminate, the chance that a person is a carrier for a particular disorder.

Please refer to the donor's Horizon Natera expanded carrier test report for more information on the testing completed and the donor's results.

**Please also see the Health Problems List for a summary of the information that this donor has provided to us regarding personal and family medical history.**

Sincerely,
Janine Mash
LCGC Certified Genetic Counselor
San Francisco Genetic Counseling



**2115 MILVIA ST BERKLEY, CA 94704 THESPERMBANKOFCA.ORG 510.841.1858**

5419

| Patient Information | |
|---|---|
| Patient Name: | |
| Date Of Birth: | |
| Gender: | Male |
| Ethnicity: | Northern European Caucasian |
| Patient ID: | N/A |
| Medical Record #: | N/A |
| Collection Kit: | 27368882-2-C |
| Accession ID: | N/A |
| Case File ID: | |
| Partner Information: | , |

| Test Information | |
|---|---|
| Ordering Physician: | Dr. Marcelle Cedars, MD (C41906) |
| Clinic Information: | UCSF |
| Phone: | 415-353-7475, 415- |
| Report Date: | 07/28/2023 |
| Sample Collected: | 07/17/2023 |
| Sample Received: | 07/18/2023 |
| 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 Glycine Encephalopathy, GLDC-Related

Positive for the pathogenic variant c.1270C>T (p.R424*) in the GLDC gene. If this individual's partner is a carrier for GLYCINE ENCEPHALOPATHY, GLDC-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 399 out of 400 diseases

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

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

**RECOMMENDATIONS**

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

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

Lyman Meng, Ph.D
Laboratory Director, Baylor Genetics

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

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





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Conceive. Deliver.

**Patient Information**
Patient Name:

Date Of Birth:
Case File ID:

**Test Information**
Ordering Physician: Dr. Marcelle Cedars, MD (C41906)
Clinic Information: UCSF

Report Date: 07/28/2023

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# GLYCINE ENCEPHALOPATHY, GLDC-RELATED

**Understanding Your Horizon Carrier Screen Results**

## What is Glycine Encephalopathy, GLDC-Related?

Glycine Encephalopathy, GLDC-Related, also known as nonketotic hyperglycinemia (NKH), is an inherited disorder that causes damage to the brain and nervous system. Lack of a certain enzyme in the body leads to a toxic buildup in the body of a building block of protein called glycine. Affected individuals usually have symptoms shortly after birth including extreme tiredness, feeding problems, weak muscle tone, jerking movements, and breathing problems that worsen and become life-threatening. Many affected children die in infancy. Children who survive with Glycine Encephalopathy, GLDC-Related have intellectual disability, seizures, and abnormal movements. Affected males may have greater chance of survival than affected females. Some affected individuals have a milder disease with symptoms that begin in childhood or adulthood. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

## What causes Glycine Encephalopathy, GLDC-Related?

Glycine Encephalopathy, GLDC-Related is caused by a gene change, or mutation, in both copies of the GLDC gene pair. These mutations cause the genes to not work properly or not work at all. The job of the GLDC genes is to breakdown glycine (a building block of protein) in the body. When both copies of this gene do not work correctly, it leads to a buildup of glycine in the body, especially the brain, and causes the symptoms described above. Glycine Encephalopathy, GLDC-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 GLDC gene to have a child with Glycine Encephalopathy, GLDC-Related. People who are carriers for Glycine Encephalopathy, GLDC- Related are usually healthy and do not have symptoms nor do they have the disorder themselves. Usually a child inherits two copies of each gene, one copy from the mother and one copy from the father. If the mother and father are both carriers for Glycine Encephalopathy, GLDC-Related, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their GLDC gene mutations to the child, who will then have this condition. Individuals found to carry more than one mutation for Glycine Encephalopathy, GLDC-Related should discuss their risk for having an affected child and any potential effects to their own health with their health care provider. There are other forms of Glycine Encephalopathy, each caused by mutations in different genes. A person who is a carrier for Glycine Encephalopathy, GLDC-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). 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 Glycine Encephalopathy, GLDC-Related ordered by a health care professional. If your partner is not found to be a carrier for Glycine Encephalopathy, GLDC-Related, your risk of having a child with this condition is greatly reduced. Couples at risk of having a baby with Glycine Encephalopathy, GLDC-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 Glycine Encephalopathy, GLDC- Related ordered by a health care professional. If your partner is found to be a carrier for Glycine Encephalopathy, GLDC-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 Glycine Encephalopathy, GLDC-Related
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for Glycine Encephalopathy, GLDC-Related
- Adoption or use of a sperm or egg donor who is not a carrier for Glycine Encephalopathy, GLDC- Related

## What resources are available?

- Genetics Home Reference: http://ghr.nlm.nih.gov/condition/glycine-encephalopathy
- 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:
Case File ID:

**Test Information**
Ordering Physician: Dr. Marcelle Cedars, MD (C41906)
Clinic Information: UCSF

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## DISEASES SCREENED

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

### Autosomal Recessive

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

3
3-BETA-HYDROXYSTEROID DEHYDROGENASE TYPE II DEFICIENCY (*HSD3B2*) **negative**
3-HYDROXY-3-METHYLGLUTARYL-COENZYME A LYASE DEFICIENCY (*HMGCL*) **negative**
3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (*HADH*) **negative**
3-METHYLCROTONYL-CoA CARBOXYLASE 1 DEFICIENCY (*MCCC1*) **negative**
3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY (*MCCC2*) **negative**
3-PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY (*PHGDH*) **negative**

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

A
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**
ALPHA-1 ANTITRYPSIN DEFICIENCY (*SERPINA1*) **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**
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 SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (*SACS*) **negative**

B
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, 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**
BATTEN DISEASE, CLN3-RELATED (*CLN3*) **negative**
BERNARD-SOULIER SYNDROME, TYPE A1 (*GP1BA*) **negative**
BERNARD-SOULIER SYNDROME, TYPE C (GP9) **negative**
BETA-HEMOGLOBINOPATHIES (*HBB*) **negative**
BETA-KETOTHIOLASE DEFICIENCY (*ACAT1*) **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**

C
CANAVAN DISEASE (*ASPA*) **negative**
CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (*CPS1*) **negative**
CARNITINE DEFICIENCY (*SLC22A5*) **negative**
CARNITINE PALMITOYLTRANSFERASE I A DEFICIENCY (*CPT1A*) **negative**
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (*CPT2*) **negative**
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (*SLC25A20*) **negative**
CARPENTER SYNDROME (*RAB23*) **negative**
CARTILAGE-HAIR HYPOPLASIA (*RMRP*) **negative**
CEREBROTENDINOUS XANTHOMATOSIS (*CYP27A1*) **negative**
CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (*NDRG1*) **negative**
CHEDIAK-HIGASHI SYNDROME (*LYST*) **negative**
CHOREOACANTHOCYTOSIS (*VPS13A*) **negative**
CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (*CYBA*) **negative**
CILIOPATHIES, RPGRIP1L-RELATED (*RPGRIP1L*) **negative**
CITRIN DEFICIENCY (*SLC25A13*) **negative**
CITRULLINEMIA, TYPE 1 (*ASS1*) **negative**
CLN10 DISEASE (*CTSD*) **negative**
COHEN SYNDROME (*VPS13B*) **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-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 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 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, HAX1-RELATED (*HAX1*) **negative**
CONGENITAL NEUTROPENIA, VPS45-RELATED (*VPS45*) **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**

D
D-BIFUNCTIONAL PROTEIN DEFICIENCY (*HSD17B4*) **negative**
DEAFNESS, AUTOSOMAL RECESSIVE 77 (*LOXHD1*) **negative**
DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY (*DPYD*) **negative**
DONNAI-BARROW SYNDROME (*LRP2*) **negative**
DYSKERATOSIS CONGENITA, RTEL1-RELATED (*RTEL1*) **negative**
DYSTROPHIC EPIDERMOLYSIS BULLOSA, COL7A1-Related (*COL7A1*) **negative**

E
EHLERS-DANLOS SYNDROME, CLASSIC-LIKE, TNXB-RELATED (*TNXB*) **negative**

**Patient Information**
Patient Name:

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horizon™ natera carrier screen

E
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**
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 HYPERCHOLESTEROLEMIA, LDLRAP1-RELATED (*LDLRAP1*) **negative**
FAMILIAL HYPERCHOLESTEROLEMIA, LDLR-RELATED (*LDLR*) **negative**
FAMILIAL HYPERINSULINISM, ABCC8-RELATED (*ABCC8*) **negative**
FAMILIAL MEDITERRANEAN FEVER (*MEFV*) **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 L (*FANCL*) **negative**
FARBER LIPOGRANULOMATOSIS (*ASAH1*) **negative**
FRASER SYNDROME 3, GRIP1-RELATED (*GRIP1*) **negative**
FRIEDREICH ATAXIA (*FXN*) **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**
GITELMAN SYNDROME (*SLC12A3*) **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**
GLYCINE ENCEPHALOPATHY, AMT-RELATED (*AMT*) **negative**
GLYCINE ENCEPHALOPATHY, GLDC-RELATED (*GLDC*) **see first page**
GLYCOGEN STORAGE DISEASE TYPE 5 ( McArdle Disease ) (*PYGM*) **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**
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 SPASTIC PARAPARESIS, TYPE 49 (*TECPR2*) **negative**
HERMANSKY-PUDLAK SYNDROME, AP3B1-RELATED (*AP3B1*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED (*HPS1*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS3-RELATED (*HPS3*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS4-RELATED (*HPS4*) **negative**
HOLOCARBOXYLASE SYNTHETASE DEFICIENCY (*HLCS*) **negative**
HOMOCYSTINURIA DUE TO DEFICIENCY OF MTHFR (*MTHFR*) **negative**
HOMOCYSTINURIA, CBS-RELATED (*CBS*) **negative**
HOMOCYSTINURIA, Type cblE (*MTRR*) **negative**
HYDROLETHALUS SYNDROME (*HYLS1*) **negative**
HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA ( HHH SYNDROME ) (*SLC25A15*) **negative**
HYPERPHOSPHATEMIC FAMILIAL TUMORAL CALCINOSIS, GALNT3-RELATED (*GALNT3*) **negative**
HYPOPHOSPHATASIA, ALPL-RELATED (*ALPL*) **negative**

I
INCLUSION BODY MYOPATHY 2 (*GNE*) **negative**
INFANTILE CEREBRAL AND CEREBELLAR ATROPHY (*MED17*) **negative**
INFANTILE NEPHRONOPHTHISIS (*INVS*) **negative**
INFANTILE NEUROAXONAL DYSTROPHY (*PLA2G6*) **negative**
ISOVALERIC ACIDEMIA (*IVD*) **negative**

J
JOHANSON-BLIZZARD SYNDROME (*UBR1*) **negative**
JOUBERT SYNDROME 2 / MECKEL SYNDROME 2 (*TMEM216*) **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, 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**
LEBER CONGENITAL AMAUROSIS 2 (*RPE65*) **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**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2 E (*SGCB*) **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 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**
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**
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 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**

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M
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 17 (*NDUFAF6*) **negative**
MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 2, SCO2-RELATED (*SCO2*) **negative**
MITOCHONDRIAL MYOPATHY AND SIDEROBLASTIC ANEMIA ( MLASA1 ) (*PUS1*) **negative**
MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (*HADHB*) **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 B / GM1 GANGLIOSIDOSIS (*GLB1*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE IVA (MORQUIO SYNDROME) (*GALNS*) **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**
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**
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**

O
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**
OSTEOPETROSIS, INFANTILE MALIGNANT, TCIRG1-RELATED (*TCIRG1*) **negative**

P
PENDRED SYNDROME (*SLC26A4*) **negative**
PERLMAN SYNDROME (*DIS3L2*) **negative**
PHENYLKETONURIA (*PAH*) **negative**
PITUITARY HORMONE DEFICIENCY, COMBINED 3 (*LHX3*) **negative**
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, 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 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**
PROLIDASE DEFICIENCY (*PEPD*) **negative**
PROPIONIC ACIDEMIA, PCCA-RELATED (*PCCA*) **negative**
PROPIONIC ACIDEMIA, PCCB-RELATED (*PCCB*) **negative**
PSEUDOCHOLINESTERASE DEFICIENCY (*BCHE*) **negative**
PSEUDOXANTHOMA ELASTICUM (*ABCC6*) **negative**
PYCNODYSOSTOSIS (*CTSK*) **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 59 (*DHDDS*) **negative**
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1 (*PEX7*) **negative**
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2 (*GNPAT*) **negative**
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3 (*AGPS*) **negative**
ROBERTS SYNDROME (*ESCO2*) **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**
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**
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.**
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 10 (*ANO10*) **negative**
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12 (*WWOX*) **negative**
SPONDYLOTHORACIC DYSOSTOSIS, MESP2-Related (*MESP2*) **negative**
STEEL SYNDROME (*COL27A1*) **negative**
STEROID-RESISTANT NEPHROTIC SYNDROME (*NPHS2*) **negative**
STUVE-WIEDEMANN SYNDROME (*LIFR*) **negative**
SURFACTANT DYSFUNCTION, ABCA3-RELATED (*ABCA3*) **negative**

T
TAY-SACHS DISEASE (*HEXA*) **negative**
TRICHOHEPATOENTERIC SYNDROME, TTC37-RELATED (*TTC37*) **negative**
TRICHOTHIODYSTROPHY 1 / XERODERMA PIGMENTOSUM, GROUP D (*ERCC2*) **negative**
TRIMETHYLAMINURIA (*FMO3*) **negative**
TRIPLE A SYNDROME (*AAAS*) **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**

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V
VERY LONG-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (*ACADVL*) **negative**
VITAMIN D-DEPENDENT RICKETS, TYPE 1A (*CYP27B1*) **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**
WERNER SYNDROME (*WRN*) **negative**
WILSON DISEASE (*ATP7B*) **negative**
WOLCOTT-RALLISON SYNDROME (*EIF2AK3*) **negative**
WOLMAN DISEASE (*LIPA*) **negative**

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

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

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## 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. If RPGR is included in the panel, exon 15 (also known as ORF15) is enriched with PCR amplification. 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. 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 provide detection of certain variants or portions of certain genes due to local sequence characteristics, high/low genomic complexity, or the presence of closely related pseudogenes. 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, duplication analysis will only be performed when c.955C>T (p.Q319*) is detected. Sequencing and CNV analysis may have reduced sensitivity, if variants result from complex gene conversion events. This test does not detect individuals with CYP21A2 gene duplication on one chromosome and CYP21A2 gene deletion on the other chromosome.

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

### 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. This analysis does not detect deletions or point mutations, which comprise about four percent of the FXN pathogenic variants.

### Friedreich Ataxia Repeat Categories

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

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## Spinal Muscular Atrophy (SMN1)

The total combined copy number of SMN1 and SMN2 exon 7 is quantified based on NGS read depth. The ratio of SMN1 to SMN2 is calculated based on the read depth of a single nucleotide that distinguishes these two genes in exon 7. In addition to copy number analysis, testing for the presence or absence of a single nucleotide polymorphism (g.27134T>G in intron 7 of SMN1) associated with the presence of a SMN1 duplication allele is performed using NGS.

| Ethnicity | Two SMN1 copies carrier risk before g.27134T>G testing | Carrier risk after g.27134T>G testing | |
|---|---|---|---|
| | | g.27134T>G ABSENT | g.27134T>G PRESENT |
| Caucasian | 1 in 632 | 1 in 769 | 1 in 29 |
| Ashkenazi Jewish | 1 in 350 | 1 in 580 | LIKELY CARRIER |
| Asian | 1 in 628 | 1 in 702 | LIKELY CARRIER |
| African-American | 1 in 121 | 1 in 396 | 1 in 34 |
| Hispanic | 1 in 1061 | 1 in 1762 | 1 in 140 |

## Variant Classification

Only pathogenic or likely pathogenic variants are reported. Other variants including benign variants, likely benign variants, variants of uncertain significance, or inconclusive variants identified during this analysis may be reported in certain circumstances. Our laboratory's variant classification criteria are based on the ACMG and internal guidelines and our current understanding of the specific genes. This interpretation may change over time as more information about a gene and/or variant becomes available. Natera and its lab partner(s) may reclassify variants at certain intervals but may not release updated reports without a specific request made to Natera by the ordering provider. Natera may disclose incidental findings if deemed clinically pertinent to the test performed.

## Negative Results

A negative carrier screening result reduces the risk for a patient to be a carrier of a specific disease but does not completely rule out carrier status. Please visit https://www.natera.com/panel-option/h-all/ for a table of carrier rates, detection rates, residual risks and promised variants/exons per gene. Carrier rates before and after testing vary by ethnicity and assume a negative family history for each disease screened and the absence of clinical symptoms in the patient. Any patient with a family history for a specific genetic disease will have a higher carrier risk prior to testing and, if the disease-causing mutation in their family is not included on the test, their carrier risk would remain unchanged. Genetic counseling is recommended for patients with a family history of genetic disease so that risk figures based on actual family history can be determined and discussed along with potential implications for reproduction. Horizon carrier screening has been developed to identify the reproductive risks for monogenic inherited conditions. Even when one or both members of a couple screen negative for pathogenic variants in a specific gene, the disease risk for their offspring is not zero. There is still a low risk for the condition in their offspring due to a number of different mechanisms that are not detected by Horizon including, but not limited to, pathogenic variant(s) in the tested gene or in a different gene not included on Horizon, pathogenic variant(s) in an upstream regulator, uniparental disomy, de novo mutation(s), or digenic or polygenic inheritance.

## Additional Comments

These analyses generally provide highly accurate information regarding the patient's carrier status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.