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 6172

I, the undersigned recipient, understand that this donor has tested **POSITIVE** as a carrier for the following condition(s). More information regarding these conditions can be found in his Expanded Carrier Screening results.

- **Mucopolysaccharidosis, Type I (IDUA)**
- **TMPRSS3-related Nonsyndromic Hearing Loss**
- **Phenylketonuria (PAH)**

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 Mucopolysaccharidosis, Type I (IDUA), TMPRSS3-related Nonsyndromic Hearing Loss and Phenylketonuria (PAH).**

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 Mucopolysaccharidosis, Type I (IDUA), TMPRSS3-related Nonsyndromic Hearing Loss and Phenylketonuria (PAH). 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
2115 Milvia Street, Berkeley Ca 94704 Phone 510.841.1858 Fax: 510.841.0332 Email: staff@tsbca.org

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: 5/8/2024

Reproductive Technologies, Inc.

THE SPERM BANK OF CALIFORNIA

EXPANDED CARRIER SCREENING RESULTS DONOR 6172

Expanded carrier screening for 524 autosomal recessive conditions was completed by Natera and reported on 4/23/2024.

The results were **POSITIVE** for **Mucopolysaccharidosis, Type I (IDUA), TMPRSS3-related Nonsyndromic Hearing Loss and Phenylketonuria (PAH)**. Donor 6172 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 and Middle Eastern ancestry)
Mucopolysaccharidosis, Type I (IDUA)	POSITIVE	n/a
TMPRSS3-related Nonsyndromic Hearing Loss	POSITIVE	n/a
Phenylketonuria (PAH)	POSITIVE	n/a
Cystic Fibrosis	Negative	1 in 2401
Spinal Muscular Atrophy	Negative: 2 copies exon 7 c.*3+80T>G variant not detected	1 in 769
HBB Hemoglobinopathies & Thalassemia	Negative	1 in 401
Alpha Thalassemia	Negative	1 in 2401

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 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

Patient Information	
Patient Name:	6172 DONOR
Date Of Birth:	
Gender:	Male
Ethnicity:	Other
Patient ID:	CONNECT-SO-273228
Medical Record #:	N/A
Collection Kit:	31707737-2-C
Accession ID:	12100151
Case File ID:	

Test Information	
Ordering Physician:	Lorraine Bonner, MD
Clinic Information:	The Sperm Bank of California
Phone:	510-841-1858
Report Date:	04/15/2024
Sample Collected:	04/03/2024
Sample Received:	04/04/2024
Sample Type:	Saliva

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 Mucopolysaccharidosis, Type I (Hurler Syndrome)
Positive for the pathogenic variant c.1205G>A (p.W402*) in the IDUA gene. If this individual's partner is a carrier for MUCOPOLYSACCHARIDOSIS, TYPE I (HURLER SYNDROME), their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

CARRIER for Nonsyndromic Hearing Loss, TMPRSS3-Related
Positive for the pathogenic variant c.916G>A (p.A306T) in the TMPRSS3 gene. If this individual's partner is a carrier for NONSYNDROMIC HEARING LOSS, TMPRSS3-RELATED, their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

CARRIER for Phenylketonuria
Positive for the pathogenic variant c.964G>A (p.A322T) in the PAH gene. If this individual's partner is a carrier for PHENYLKETONURIA, 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 521 out of 524 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

Linyan 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

natera® Conceive. Deliver.

Patient Information		Test Information	
Patient Name:	6172 DONOR	Ordering Physician:	Lorraine Bonner, MD
		Clinic Information:	The Sperm Bank of California
Date Of Birth:			
Case File ID:	12100151		
		Report Date:	04/15/2024

MUCOPOLYSACCHARIDOSIS, TYPE I (HURLER SYNDROME)

Understanding Your Horizon Carrier Screen Results

What is Mucopolysaccharidosis, Type I?

Mucopolysaccharidosis (MPS), Type I, also known as Hurler syndrome, is an inherited disorder that causes toxic buildup of certain types of sugars, called glycosaminoglycans, in the body. There are mild and severe forms of MPS, Type I. Most children with MPS, Type I have symptoms in the first years of life including progressive intellectual disability, large head size, coarse facial features, heart problems, bone problems, short stature, enlarged liver and spleen, frequent infections, vision problems, and breathing problems. Without treatment, children with this form of MPS, Type I usually do not survive past childhood. People with milder forms of MPS, Type I usually live into adulthood and have milder intellectual disability. Treatment may include enzyme replacement therapy. In some cases, affected individuals have been treated with stem cell transplantation from cord blood or bone marrow. Couples at risk of having an affected child may consider cord blood banking, as siblings have a higher chance of being a match for stem cell transplantation than a non-related individual. More information can be found at: https://parentsguidecordblood.org/en. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Mucopolysaccharidosis, Type I?

MPS, Type I is caused by a gene change, or mutation, in both copies of the IDUA gene pair. These mutations cause the genes to not work properly or not work at all. The function of the IDUA genes is to create an enzyme which breaks down long chain sugar molecules and clears them from the body. When both copies of this gene do not work correctly, it causes buildup of certain sugars over time causing cell damage in many organs. This leads to the symptoms described above. MPS, Type I 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 IDUA gene to have a child with MPS, Type I. People who are carriers for MPS, Type I are usually healthy and do not have symptoms nor do they have MPS, Type I themselves. Usually a child inherits two copies of each gene, one copy from the mother and one copy from the father. If the mother and father are both carriers for MPS, Type I there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their IDUA gene mutations to the child, who would then have MPS, Type I. Individuals found to carry more than one mutation for MPS, Type I should discuss their risk for having an affected child with their health care provider. There are many other types of Mucopolysaccharidosis (MPS), each caused by mutations in different genes. A carrier for MPS, Type I is not likely to be at increased risk for having children with the other forms of MPS.

What can I do next?

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website (www.nsgc.org). Your siblings and other relatives are at increased risk to also have this mutation. You are encouraged to inform your family members of your test results as they may wish to consider being tested themselves. If you are pregnant, your partner can have carrier screening for MPS, Type I ordered by a health care professional. If your partner is not found to be a carrier for MPS, Type I your risk of having a child with MPS, Type I is greatly reduced. Couples at risk of having a baby with MPS, Type I 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 MPS, Type I ordered by a health care professional. If your partner is found to be a carrier for MPS, Type I you have several reproductive options to consider:

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

What resources are available?

- National MPS Society: http://mpssociety.org/
- Prenatal diagnosis done through CVS: http://www.marchofdimes.org/chorionic-villus-sampling.aspx
- Prenatal diagnosis done through Amniocentesis: http://www.marchofdimes.org/amniocentesis.aspx
- Preimplantation genetic diagnosis (PGD) with IVF: http://www.natera.com/spectrum

Patient Information
Patient Name:

Date Of Birth:
Case File ID:

Test Information
Ordering Physician:

Clinic Information:

Report Date:

NONSYNDROMIC HEARING LOSS, TMPRSS3-RELATED

Understanding Your Horizon Carrier Screen Results

What does being a carrier mean?

Your results show that you are a carrier of nonsyndromic hearing loss, TMPRSS3-related (also known as DFNB8/10). A carrier of a genetic condition does not have the condition. Carriers also are not certain to have a child with the condition. We are all carriers of one or more genetic conditions.

Your children are not at high risk for this condition unless your partner or donor is also a carrier of DFNB8/10. Further testing can be done to see if your partner or donor is a carrier.

What is nonsyndromic hearing loss, TMPRSS3-related (DFNB8/10)?

DFNB8/10 causes hearing loss. People with DFNB8/10 can have hearing loss starting in early childhood, before they learn to speak, or later in childhood (10-12 years). Sometimes the hearing loss gets worse over time. This type of hearing loss, called sensorineural, can often be helped by hearing aids or implants. This condition does not cause any health problems other than hearing loss.[1,2,3]

Clinical trials involving potential new treatments for this condition could be available (see clinicaltrials.gov).

What causes nonsyndromic hearing loss, TMPRSS3-related (DFNB8/10)?

DFNB8/10 is caused by changes, or variants, in the TMPRSS3 gene. These changes make the gene not work properly. Genes are a set of instructions inside the cells of our bodies that tell our bodies how to grow and function. Everyone has two copies of the TMPRSS3 gene. Carriers of DFNB8/10 have one working copy and one non-working copy of the gene. People with DFNB8/10 have no working copies of the gene.

DFNB8/10 is usually passed down, or inherited, from both genetic parents. We inherit one copy of the TMPRSS3 gene from each of our genetic parents. When both genetic parents are carriers, each child has a 1 in 4 (25%) chance of inheriting two non-working genes and having DFNB8/10. Each child also has a 1 in 2 (50%) chance of being a carrier of DFNB8/10 and a 1 in 4 (25%) chance of inheriting two working copies of the gene. This type of inheritance is called autosomal recessive inheritance.

Will my children have nonsyndromic hearing loss, TMPRSS3-related (DFNB8/10)?

If your partner or donor also has a non-working copy of the TMPRSS3 gene, your children could have DFNB8/10. Each child you have together would have a 1 in 4 (25%) chance of having DFNB8/10. Each child you have together would also have a 3 in 4 (75%) chance of **not** having the condition.

If your partner or donor has TMPRSS3 carrier screening and no variants are found, the chance that your children would have DFNB8/10 is very low. No further testing would usually be needed for you, your partner or donor, or your children related to DFNB8/10.

What can I do next?

If you want to know if your children are at risk for DFNB8/10, your partner or donor would need to have TMPRSS3 carrier screening. If you have questions about this testing, please ask your healthcare provider or use the resources below. Many people find it helpful to speak with a genetic counselor.

If your partner or donor is found to be a DFNB8/10 carrier, your children would be at risk for having DFNB8/10.

If you or your partner or surrogate are currently pregnant, tests called CVS (chorionic villus sampling) and amniocentesis can be done during pregnancy to find out if a baby has DFNB8/10. These tests both have a small risk of miscarriage. Babies can also be tested for DFNB8/10 after birth instead.

If you or your partner or surrogate are not yet pregnant, you could have these options:

- natural pregnancy and testing the baby after birth for DFNB8/10;
- preimplantation genetic testing (PGT-M) with in vitro fertilization (IVF) to test embryos for DFNB8/10;
- adoption; or
- use of a sperm or egg donor who had no variants found in TMPRSS3 carrier screening.

Where can I find more information?

- Hearing Health Foundation hearinghealthfoundation.org
- Hearing Loss Association of America hearingloss.org/
- CVS marchofdimes.org/chorionic-villus-sampling
- Amniocentesis marchofdimes.org/pregnancy/amniocentesis
- PGT-M natera.com/womens-health/spectrum-preimplantation-genetics

What does this mean for my family?

You likely got (inherited) this non-working gene from one of your genetic parents. Your genetic siblings and other family members could also carry it. You should tell your family members about your test results so they can decide if they want carrier screening for DFNB8/10.

References

1. Online Mendelian Inheritance in Man, OMIM®. Johns Hopkins University, Baltimore, MD. DEAFNESS, AUTOSOMAL RECESSIVE 8; DFNB8. MIM Number:

601072: 06/01/2016: Available from: https://www.omim.org/entry/601072. Accessed 01/24.
2. Weegerink NJ et al. Genotype-phenotype correlation in DFNB8/10 families with TMPRSS3 mutations. J Assoc Res Otolaryngol. 2011 Dec;12(6):753-66. doi: 10.1007/s10162-011-0282-3. Epub 2011 Jul 23. PMID: 21786053; PMCID: PMC3214237.
3. Gao X et al. Identification of TMPRSS3 as a Significant Contributor to Autosomal Recessive Hearing Loss in the Chinese Population. Neural Plast. 2017;2017:3192090. doi: 10.1155/2017/3192090. Epub 2017 Jun 13. PMID: 28695016; PMCID: PMC5485344.

Patient Information
Patient Name:

Date Of Birth:
Case File ID:

Test Information
Ordering Physician:

Clinic Information:

Report Date:

horizon™ natera carrier screen

PHENYLKETONURIA

Understanding Your Horizon Carrier Screen Results

What is Phenylketonuria?

Phenylketonuria (PKU) is an inherited disorder in which the body is unable to break down an amino acid (building block of protein) called phenylalanine. Phenylalanine is found in most foods that contain protein including meat, fish, dairy, eggs, beans, and nuts. When toxic levels of phenylalanine buildup in the body it causes problems for the brain, nervous system, and other parts of the body. If the condition is not treated, children with PKU develop intellectual disability, developmental delay, seizures, skin problems, and psychiatric problems. Lifelong treatment with a diet low in phenylalanine and special supplements is typically needed to treat PKU. With treatment people with PKU can lead healthy lives. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov). Other forms of Phenylketonuria called variant PKU and non-PKU hyperphenylalaninemia can be less severe and have a lower risk for brain and health problems. Some people with very mild cases may not need treatment with a low phenylalanine diet.

What causes Phenylketonuria?

PKU is caused by a gene change, or mutation, in both copies of the PAH gene pair. These mutations cause the genes to not work properly or not work at all. Normal function of the PAH genes is important for breaking down phenylalanine from foods in the diet. When both copies of the PAH gene do not work correctly, it leads to the symptoms described above. PKU 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 PAH gene to have a child with PKU. People who are carriers for PKU are usually healthy and do not have symptoms nor do they have PKU 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 PKU, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their PAH gene mutations to the child, who will then have this condition. Individuals found to carry more than one mutation for Phenylketonuria should discuss their risk for having an affected child, and any potential effects to their own health, with their health care provider.

What can I do next?

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website (www.nsgc.org). Your siblings and other relatives are at increased risk to also have this mutation. You are encouraged to inform your family members of your test results as they may wish to consider being tested themselves. If you are pregnant, your partner can have carrier screening for PKU ordered by a health care professional. If your partner is not found to be a carrier for PKU your risk of having an affected child is greatly reduced. Couples at risk of having a baby with PKU 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. Although PKU is screened for as part of the newborn screening program in all U.S. states, babies at 25% risk for this condition may need diagnostic testing in addition to newborn screening. If you are not yet pregnant, your partner can have carrier screening for PKU ordered by a health care professional. If your partner is found to be a carrier for PKU you have several reproductive options to consider:

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

What resources are available?

- Baby's First Test: https://www.babysfirsttest.org/newborn-screening/conditions/classic- phenylketonuria-pku
- Genetics Home Reference: https://ghr.nlm.nih.gov/condition/phenylketonuria
- 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:

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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**

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**
ACUTE INFANTILE LIVER FAILURE, TRMU-RELATED (*TRMU*) **negative**
ACYL-COA OXIDASE I DEFICIENCY (*ACOX1*) **negative**
AICARDI-GOUTIÈRES SYNDROME (*SAMHD1*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2A-RELATED (*RNASEH2A*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2B-RELATED (*RNASEH2B*) **negative**
AICARDI-GOUTIERES SYNDROME, RNASEH2C-RELATED (*RNASEH2C*) **negative**
AICARDI-GOUTIÈRES SYNDROME, TREX1-RELATED (*TREX1*) **negative**
ALKAPTONURIA (*HGD*) **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**
ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (*GATM*) **negative**
ARGININEMIA (*ARG1*) **negative**
ARGININOSUCCINATE LYASE DEFICIENCY (*ASL*) **negative**
AROMATASE DEFICIENCY (*CYP19A1*) **negative**
ASPARAGINE SYNTHETASE DEFICIENCY (*ASNS*) **negative**
ASPARTYLGLYCOSAMINURIA (*AGA*) **negative**
ATAXIA WITH VITAMIN E DEFICIENCY (*TTPA*) **negative**
ATAXIA-TELANGIECTASIA (*ATM*) **negative**
ATAXIA-TELANGIECTASIA-LIKE DISORDER 1 (*MRE11*) **negative**
ATRANSFERRINEMIA (*TF*) **negative**
AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (*SLC35A3*) **negative**
AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (*AIRE*) **negative**
AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (ARCI), SLC27A4-RELATED (*SLC27A4*) **negative**
AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (*SACS*) **negative**

B
BARDET-BIEDL SYNDROME, ARL6-RELATED (*ARL6*) **negative**
BARDET-BIEDL SYNDROME, BBS10-RELATED (*BBS10*) **negative**
BARDET-BIEDL SYNDROME, BBS12-RELATED (*BBS12*) **negative**
BARDET-BIEDL SYNDROME, BBS1-RELATED (*BBS1*) **negative**
BARDET-BIEDL SYNDROME, BBS2-RELATED (*BBS2*) **negative**
BARDET-BIEDL SYNDROME, BBS4-RELATED (*BBS4*) **negative**
BARDET-BIEDL SYNDROME, BBS5-RELATED (*BBS5*) **negative**
BARDET-BIEDL SYNDROME, BBS7-RELATED (*BBS7*) **negative**
BARDET-BIEDL SYNDROME, BBS9-RELATED (*BBS9*) **negative**
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**
BERNARD-SOULIER SYNDROME, TYPE A1 (*GP1BA*) **negative**
BERNARD-SOULIER SYNDROME, TYPE C (*GP9*) **negative**
BETA-HEMOGLOBINOPATHIES (*HBB*) **negative**
BETA-KETOTHIOLASE DEFICIENCY (*ACAT1*) **negative**
BETA-MANNOSIDOSIS (*MANBA*) **negative**
BILATERAL FRONTOPARIETAL POLYMICROGYRIA (*GPR56*) **negative**
BIOTINIDASE DEFICIENCY (*BTD*) **negative**
BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBGD) (*SLC19A3*) **negative**
BLOOM SYNDROME (*BLM*) **negative**
BRITTLE CORNEA SYNDROME 1 (*ZNF469*) **negative**
BRITTLE CORNEA SYNDROME 2 (*PRDM5*) **negative**

C
CANAVAN DISEASE (*ASPA*) **negative**
CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (*CPS1*) **negative**
CARNITINE DEFICIENCY (*SLC22A5*) **negative**
CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY (*CPT1A*) **negative**
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (*CPT2*) **negative**
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (*SLC25A20*) **negative**
CARPENTER SYNDROME (*RAB23*) **negative**
CARTILAGE-HAIR HYPOPLASIA (*RMRP*) **negative**
CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (*CASQ2*) **negative**
CD59-MEDIATED HEMOLYTIC ANEMIA (*CD59*) **negative**
CEP152-RELATED MICROCEPHALY (*CEP152*) **negative**
CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA (CEDNIK) SYNDROME (*SNAP29*) **negative**
CEREBROTENDINOUS XANTHOMATOSIS (*CYP27A1*) **negative**
CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE C (*PLEKHG5*) **negative**
CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (*NDRG1*) **negative**
CHEDIAK-HIGASHI SYNDROME (*LYST*) **negative**
CHOREOACANTHOCYTOSIS (*VPS13A*) **negative**
CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (*CYBA*) **negative**
CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (*NCF2*) **negative**
CILIOPATHIES, RPGRIP1L-RELATED (*RPGRIP1L*) **negative**
CITRIN DEFICIENCY (*SLC25A13*) **negative**
CITRULLINEMIA, TYPE 1 (*ASS1*) **negative**
CLN10 DISEASE (*CTSD*) **negative**
COHEN SYNDROME (*VPS13B*) **negative**
COL11A2-RELATED CONDITIONS (*COL11A2*) **negative**
COMBINED MALONIC AND METHYLMALONIC ACIDURIA (*ACSF3*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (*GFM1*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (*TSFM*) **negative**
COMBINED PITUITARY HORMONE DEFICIENCY 1 (*POU1F1*) **negative**
COMBINED PITUITARY HORMONE DEFICIENCY-2 (*PROP1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (*CYP11B1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (*CYP17A1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (*CYP21A2*) **negative**
CONGENITAL ADRENAL INSUFFICIENCY, CYP11A1-RELATED (*CYP11A1*) **negative**
CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (*MPL*) **negative**
CONGENITAL CHRONIC DIARRHEA (*DGAT1*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, ALG1-RELATED (*ALG1*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-Related (*PMM2*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (*MPI*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (*ALG6*) **negative**
CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (*SEC23B*) **negative**
CONGENITAL FINNISH NEPHROSIS (*NPHS1*) **negative**
CONGENITAL HYDROCEPHALUS 1 (*CCDC88C*) **negative**
CONGENITAL HYPERINSULINISM, KCNJ11-Related (*KCNJ11*) **negative**
CONGENITAL 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, DOK7-RELATED (*DOK7*) **negative**
CONGENITAL MYASTHENIC SYNDROME, RAPSN-RELATED (*RAPSN*) **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**

Patient Information
Patient Name:

Date Of Birth:
Case File ID:

Test Information
Ordering Physician:

Clinic Information:

Report Date:

horizon™ natera carrier screen

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, RTEL1-RELATED (*RTEL1*) **negative**
DYSTROPHIC EPIDERMOLYSIS BULLOSA, COL7A1-Related (*COL7A1*) **negative**

E
EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY, CAD-RELATED (*CAD*) **negative**
EHLERS-DANLOS SYNDROME TYPE VI (*PLOD1*) **negative**
EHLERS-DANLOS SYNDROME, 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
F2-RELATED CONDITIONS (*F2*) **negative**
F5-RELATED CONDITIONS (*F5*) **negative**
FACTOR XI DEFICIENCY (*F11*) **negative**
FAMILIAL DYSAUTONOMIA (*IKBKAP*) **negative**
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, PRF1-RELATED (*PRF1*) **negative**
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STX11-RELATED (*STX11*) **negative**
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STXBP2-RELATED (*STXBP2*) **negative**
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, UNC13D-RELATED (*UNC13D*) **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 G (*FANCG*) **negative**
FANCONI ANEMIA, GROUP I (*FANCI*) **negative**
FANCONI ANEMIA, GROUP J (*BRIP1*) **negative**
FANCONI ANEMIA, GROUP L (*FANCL*) **negative**
FOVEAL HYPOPLASIA (*SLC38A8*) **negative**
FRASER SYNDROME 3, GRIP1-RELATED (*GRIP1*) **negative**
FRASER SYNDROME, FRAS1-RELATED (*FRAS1*) **negative**
FRASER SYNDROME, FREM2-RELATED (*FREM2*) **negative**
FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY (*FBP1*) **negative**
FUCOSIDOSIS, FUCA1-RELATED (*FUCA1*) **negative**
FUMARASE DEFICIENCY (*FH*) **negative**

G
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 1 (*HFE*) **negative**
HEREDITARY HEMOCHROMATOSIS TYPE 2B (*HAMP*) **negative**
HEREDITARY SPASTIC PARAPARESIS, TYPE 49 (*TECPR2*) **negative**
HEREDITARY SPASTIC PARAPLEGIA, CYP7B1-RELATED (*CYP7B1*) **negative**
HERMANSKY-PUDLAK SYNDROME, BLOC1S3-RELATED (*BLOC1S3*) **negative**
HERMANSKY-PUDLAK SYNDROME, BLOC1S6-RELATED (*BLOC1S6*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED (*HPS1*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS3-RELATED (*HPS3*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS4-RELATED (*HPS4*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS5-RELATED (*HPS5*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS6-RELATED (*HPS6*) **negative**
HOLOCARBOXYLASE SYNTHETASE DEFICIENCY (*HLCS*) **negative**
HOMOCYSTINURIA AND MEGALOBLASTIC ANEMIA TYPE CBLG (*MTR*) **negative**
HOMOCYSTINURIA DUE TO DEFICIENCY OF MTHFR (*MTHFR*) **negative**
HOMOCYSTINURIA, CBS-RELATED (*CBS*) **negative**
HOMOCYSTINURIA, Type cblE (*MTRR*) **negative**
HYDROLETHALUS SYNDROME (*HYLS1*) **negative**
HYPER-IGM IMMUNODEFICIENCY (*CD40*) **negative**
HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA (HHH SYNDROME) (*SLC25A15*) **negative**
HYPERPHOSPHATEMIC FAMILIAL TUMORAL CALCINOSIS, GALNT3-RELATED (*GALNT3*) **negative**
HYPOMYELINATING LEUKODYSTROPHY 12 (*VPS11*) **negative**
HYPOPHOSPHATASIA, ALPL-RELATED (*ALPL*) **negative**

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

J
JOHANSON-BLIZZARD SYNDROME (*UBR1*) **negative**
JOUBERT SYNDROME 2 / MECKEL SYNDROME 2 (*TMEM216*) **negative**
JOUBERT SYNDROME AND RELATED DISORDERS (JSRD), TMEM67-RELATED (*TMEM67*) **negative**
JOUBERT SYNDROME, AHI1-RELATED (*AHI1*) **negative**
JOUBERT SYNDROME, CC2D2A-RELATED/COACH SYNDROME (*CC2D2A*) **negative**
JUNCTIONAL EPIDERMOLYSIS BULLOSA, COL17A1-RELATED (*COL17A1*) **negative**
JUNCTIONAL EPIDERMOLYSIS BULLOSA, ITGA6-RELATED (*ITGA6*) **negative**
JUNCTIONAL EPIDERMOLYSIS BULLOSA, ITGB4-RELATED (*ITGB4*) **negative**
JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMB3-RELATED (*LAMB3*) **negative**
JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMC2-RELATED (*LAMC2*) **negative**
JUNCTIONAL EPIDERMOLYSIS BULLOSA/LARYNGOONYCHOCUTANEOUS SYNDROME, LAMA3-RELATED (*LAMA3*) **negative**

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

L
LAMELLAR ICHTHYOSIS, TYPE 1 (*TGM1*) **negative**
LARON SYNDROME (*GHR*) **negative**
LEBER CONGENITAL AMAUROSIS 2 (*RPE65*) **negative**
LEBER CONGENITAL AMAUROSIS TYPE AIPL1 (*AIPL1*) **negative**
LEBER CONGENITAL AMAUROSIS TYPE GUCY2D (*GUCY2D*) **negative**
LEBER CONGENITAL AMAUROSIS TYPE TULP1 (*TULP1*) **negative**
LEBER CONGENITAL AMAUROSIS, 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**

L
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B3-RELATED (*EIF2B3*) **negative**
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B4-RELATED (*EIF2B4*) **negative**
LIG4 SYNDROME (*LIG4*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 8 (*TRIM32*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2A (*CAPN3*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2B (*DYSF*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2C (*SGCG*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2D (*SGCA*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2E (*SGCB*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2F (*SGCD*) **negative**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2I (*FKRP*) **negative**
LIPOAMIDE DEHYDROGENASE DEFICIENCY (DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY) (*DLD*) **negative**
LIPOID ADRENAL HYPERPLASIA (*STAR*) **negative**
LIPOPROTEIN LIPASE DEFICIENCY (*LPL*) **negative**
LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (*HADHA*) **negative**
LRAT-RELATED CONDITIONS (*LRAT*) **negative**
LUNG DISEASE, IMMUNODEFICIENCY, AND CHROMOSOME BREAKAGE SYNDROME (LICS) (*NSMCE3*) **negative**
LYSINURIC PROTEIN INTOLERANCE (*SLC7A7*) **negative**

M
MALONYL-COA DECARBOXYLASE DEFICIENCY (*MLYCD*) **negative**
MAPLE SYRUP URINE DISEASE, TYPE 1A (*BCKDHA*) **negative**
MAPLE SYRUP URINE DISEASE, TYPE 1B (*BCKDHB*) **negative**
MAPLE SYRUP URINE DISEASE, TYPE 2 (*DBT*) **negative**
MCKUSICK-KAUFMAN SYNDROME (*MKKS*) **negative**
MECKEL-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 OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II (*PCNT*) **negative**
MICROPHTHALMIA / ANOPHTHALMIA, VSX2-RELATED (*VSX2*) **negative**
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, ACAD9-RELATED (*ACAD9*) **negative**
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFAF5-RELATED (*NDUFAF5*) **negative**
MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFS6-RELATED (*NDUFS6*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 1 (*NDUFS4*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 10 (*NDUFAF2*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 19 (*FOXRED1*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 3 (*NDUFS7*) **negative**
MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 4 (*NDUFV1*) **negative**
MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 2, SCO2-RELATED (*SCO2*) **negative**
MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 6 (*COX15*) **negative**
MITOCHONDRIAL DNA DEPLETION SYNDROME 2 (*TK2*) **negative**
MITOCHONDRIAL DNA DEPLETION SYNDROME 3 (*DGUOK*) **negative**
MITOCHONDRIAL MYOPATHY AND SIDEROBLASTIC ANEMIA (MLASA1) (*PUS1*) **negative**
MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (*HADHB*) **negative**
MOLYBDENUM COFACTOR DEFICIENCY TYPE B (*MOCS2*) **negative**
MOLYBDENUM COFACTOR DEFICIENCY, TYPE A (*MOCS1*) **negative**
MUCOLIPIDOSIS II/III A (*GNPTAB*) **negative**
MUCOLIPIDOSIS III GAMMA (*GNPTG*) **negative**
MUCOLIPIDOSIS, TYPE IV (*MCOLN1*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE I (HURLER SYNDROME) (*IDUA*) **see first page**
MUCOPOLYSACCHARIDOSIS, TYPE III A (SANFILIPPO A) (*SGSH*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III B (SANFILIPPO B) (*NAGLU*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III C (SANFILIPPO C) (*HGSNAT*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE III D (SANFILIPPO D) (*GNS*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE IV A (MORQUIO SYNDROME) (*GALNS*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE IV B/GM1 GANGLIOSIDOSIS (*GLB1*) **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-ACETYLGLUTAMATE SYNTHASE DEFICIENCY (*NAGS*) **negative**
NEMALINE MYOPATHY, NEB-RELATED (*NEB*) **negative**
NEPHRONOPHTHISIS 1 (*NPHP1*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, CLN5-RELATED (*CLN5*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, CLN6-RELATED (*CLN6*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, CLN8-RELATED (*CLN8*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, MFSD8-RELATED (*MFSD8*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, PPT1-RELATED (*PPT1*) **negative**
NEURONAL CEROID LIPOFUSCINOSIS, TPP1-RELATED (*TPP1*) **negative**
NGLY1-CONGENITAL DISORDER OF GLYCOSYLATION (*NGLY1*) **negative**
NIEMANN-PICK DISEASE, TYPE C1 / D (*NPC1*) **negative**
NIEMANN-PICK DISEASE, TYPE C2 (*NPC2*) **negative**
NIEMANN-PICK DISEASE, TYPES A / B (*SMPD1*) **negative**
NIJMEGEN BREAKAGE SYNDROME (*NBN*) **negative**
NON-SYNDROMIC HEARING LOSS, GJB2-RELATED (*GJB2*) **negative**
NON-SYNDROMIC HEARING LOSS, MYO15A-RELATED (*MYO15A*) **negative**
NONSYNDROMIC HEARING LOSS, OTOA-RELATED (*OTOA*) **negative**
NONSYNDROMIC HEARING LOSS, OTOF-RELATED (*OTOF*) **negative**
NONSYNDROMIC HEARING LOSS, PJVK-RELATED (*PJVK*) **negative**
NONSYNDROMIC HEARING LOSS, SYNE4-RELATED (*SYNE4*) **negative**
NONSYNDROMIC HEARING LOSS, TMC1-RELATED (*TMC1*) **negative**
NONSYNDROMIC HEARING LOSS, TMPRSS3-RELATED (*TMPRSS3*) **see first page**
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*) **negative**
OCULOCUTANEOUS ALBINISM, TYPES 1A AND 1B (*TYR*) **negative**
ODONTO-ONYCHO-DERMAL DYSPLASIA / SCHOPF-SCHULZ-PASSARGE SYNDROME (*WNT10A*) **negative**
OMENN SYNDROME, RAG2-RELATED (*RAG2*) **negative**
ORNITHINE AMINOTRANSFERASE DEFICIENCY (*OAT*) **negative**
OSTEOGENESIS IMPERFECTA TYPE VII (*CRTAP*) **negative**
OSTEOGENESIS IMPERFECTA TYPE VIII (*P3H1*) **negative**
OSTEOGENESIS IMPERFECTA TYPE XI (*FKBP10*) **negative**
OSTEOGENESIS IMPERFECTA TYPE XIII (*BMP1*) **negative**
OSTEOPETROSIS, INFANTILE MALIGNANT, TCIRG1-RELATED (*TCIRG1*) **negative**
OSTEOPETROSIS, OSTM1-RELATED (*OSTM1*) **negative**

P
PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION (*PANK2*) **negative**
PAPILLON LEFÈVRE SYNDROME (*CTSC*) **negative**
PARKINSON DISEASE 15 (*FBXO7*) **negative**
PENDRED SYNDROME (*SLC26A4*) **negative**
PGM3-CONGENITAL DISORDER OF GLYCOSYLATION (*PGM3*) **negative**
PHENYLKETONURIA (*PAH*) **see first page**
PIGN-CONGENITAL DISORDER OF GLYCOSYLATION (*PIGN*) **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, TSEN54-RELATED (*TSEN54*) **negative**
PONTOCEREBELLAR HYPOPLASIA, TYPE 1A (*VRK1*) **negative**
PONTOCEREBELLAR HYPOPLASIA, TYPE 2D (*SEPSECS*) **negative**
PONTOCEREBELLAR HYPOPLASIA, VPS53-RELATED (*VPS53*) **negative**
PRIMARY CILIARY DYSKINESIA, CCDC103-RELATED (*CCDC103*) **negative**
PRIMARY CILIARY DYSKINESIA, CCDC39-RELATED (*CCDC39*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAH11-RELATED (*DNAH11*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAH5-RELATED (*DNAH5*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAI1-RELATED (*DNAI1*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAI2-RELATED (*DNAI2*) **negative**
PRIMARY CONGENITAL GLAUCOMA/PETERS ANOMALY (*CYP1B1*) **negative**
PRIMARY HYPEROXALURIA, TYPE 1 (*AGXT*) **negative**
PRIMARY HYPEROXALURIA, TYPE 2 (*GRHPR*) **negative**
PRIMARY HYPEROXALURIA, TYPE 3 (*HOGA1*) **negative**
PRIMARY MICROCEPHALY 1, AUTOSOMAL RECESSIVE (*MCPH1*) **negative**
PROGRESSIVE EARLY-ONSET ENCEPAHLOPATHY WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM (*TBCD*) **negative**

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P
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 PSEUDORHEUMATOID DYSPLASIA (*CCN6*) **negative**
PROLIDASE DEFICIENCY (*PEPD*) **negative**
PROPIONIC ACIDEMIA, PCCA-RELATED (*PCCA*) **negative**
PROPIONIC ACIDEMIA, PCCB-RELATED (*PCCB*) **negative**
PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (PCD) DEFICIENCY (*PCBD1*) **negative**
PYCNODYSOSTOSIS (*CTSK*) **negative**
PYRIDOXAL 5'-PHOSPHATE-DEPENDENT EPILEPSY (*PNPO*) **negative**
PYRIDOXINE-DEPENDENT EPILEPSY (*ALDH7A1*) **negative**
PYRUVATE CARBOXYLASE DEFICIENCY (*PC*) **negative**
PYRUVATE DEHYDROGENASE DEFICIENCY, PDHB-RELATED (*PDHB*) **negative**

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

S
SALLA DISEASE (*SLC17A5*) **negative**
SANDHOFF DISEASE (*HEXB*) **negative**
SCHIMKE IMMUNOOSSEOUS DYSPLASIA (*SMARCAL1*) **negative**
SCHINDLER DISEASE (*NAGA*) **negative**
SEGAWA SYNDROME, TH-RELATED (*TH*) **negative**
SEPIAPTERIN REDUCTASE DEFICIENCY (*SPR*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3D-RELATED (*CD3D*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3E-RELATED (*CD3E*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), FOXN1-RELATED (*FOXN1*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), IKBKB-RELATED (*IKBKB*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), IL7R-RELATED (*IL7R*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), JAK3-RELATED (*JAK3*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), PTPRC-RELATED (*PTPRC*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), RAG1-RELATED (*RAG1*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY, ADA-Related (*ADA*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY, TYPE ATHABASKAN (*DCLRE1C*) **negative**
SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY (*DYNC2H1*) **negative**
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**
SPONDYLOCOSTAL DYSOSTOSIS 1 (*DLL3*) **negative**
SPONDYLOTHORACIC DYSOSTOSIS, MESP2-Related (*MESP2*) **negative**
STEEL SYNDROME (*COL27A1*) **negative**
STEROID-RESISTANT NEPHROTIC SYNDROME (*NPHS2*) **negative**
STUVE-WIEDEMANN SYNDROME (*LIFR*) **negative**
SURF1-RELATED CONDITIONS (*SURF1*) **negative**
SURFACTANT DYSFUNCTION, ABCA3-RELATED (*ABCA3*) **negative**

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

U
USHER SYNDROME, TYPE 1B (*MYO7A*) **negative**
USHER SYNDROME, TYPE 1C (*USH1C*) **negative**
USHER SYNDROME, TYPE 1D (*CDH23*) **negative**
USHER SYNDROME, TYPE 1F (*PCDH15*) **negative**
USHER SYNDROME, TYPE 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, 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**
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 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 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.

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.

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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.