

SPERM DONOR GENETIC TESTING SUMMARY

Donor # 8208

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

Last Updated: 06/17/2026

Donor Reported Ancestry: Chinese, Korean

Jewish Ancestry: No

Genetic Test*	Result	Comments Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Expanded Genetic Disease Carrier Screening Panel attached - 549 diseases by gene sequencing and del/dup analysis.	<p>Carrier: Deafness, Autosomal Recessive 77 (LOXHD1)</p> <p>Carrier: Hermansky-Pudlak Syndrome, HPS1-Related (HPS1)</p> <p>Carrier: Leber Congenital Amaurosis Type GUCY2D (GUCY2D)</p> <p>Carrier: Non-Syndromic Hearing Loss, GJB2-Related (GJB2)</p> <p>Carrier: Thyroid Dyshormonogenesis 6 (DUOX2)</p> <p>Negative for other genes tested.</p>	<p>Partner testing is recommended before using this donor.</p> <p>Carriers of Thyroid Dyshormonogenesis 6 (DUOX2) may have symptoms of the condition as babies that go away as they age. Please see results for further information. Genetic counseling can be considered.</p>

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

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

Patient Information

Patient Name: DONOR 8208

Date Of Birth:

Gender: Male

Patient ID: N/A

Medical Record #: 8208-

Collection Kit:

Accession ID: N/A

Case File ID:

Ethnicity: East Asian

Test Information

Ordering Physician:

Clinic Information: Fairfax Cryobank

Phone:

Report Date: 09/11/2025

Sample Collected: 08/28/2025

Sample Received: 08/29/2025

Sample Type: Blood

CARRIER SCREENING REPORT

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

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

FINAL RESULTS SUMMARY:

CARRIER for Deafness, Autosomal Recessive 77

Positive for the likely pathogenic variant c.3281A>G (p.D1094G) in the LOXHD1 gene. If this individual's partner is a carrier for DEAFNESS, AUTOSOMAL RECESSIVE 77, their chance to have a child with this condition may be as high as 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

CARRIER for Hermansky-Pudlak Syndrome, HPS1-Related

Positive for the pathogenic variant c.9C>A (p.C3*) in the HPS1 gene. If this individual's partner is a carrier for HERMANSKY-PUDLAK SYNDROME, HPS1-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 Leber Congenital Amaurosis Type GUCY2D

Positive for the likely pathogenic variant c.2927G>T (p.R976L) in the GUCY2D gene. Although most variants in this gene are associated with an autosomal recessive form of Leber congenital amaurosis type GUCY2D, some rare GUCY2D variants may cause an autosomal dominant form of the condition (PMID: 9618177, 11115851, 26298565, and 12552567). This individual's chance to have a child with Leber congenital amaurosis type GUCY2D is as high as 1 in 2 (50%). Carrier screening for this individual's partner is suggested.

CARRIER for Non-Syndromic Hearing Loss, GJB2-Related

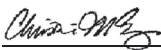
Positive for the pathogenic variant c.109G>A (p.V37I) in the GJB2 gene. This variant has been reported to have variable penetrance, and some individuals with a pathogenic variant on the opposite allele may not have hearing loss. If this individual's partner is a carrier for NON-SYNDROMIC HEARING LOSS, GJB2-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 Thyroid Dysmorphogenesis 6

Positive for the likely pathogenic variant c.514-1G>A in the DUOX2 gene. If this individual's partner is a carrier for THYROID DYSHORMONOGENESIS 6, their chance to have a child with this condition may be as high as 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

Negative for 544 out of 549 diseases

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



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



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



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Senior Laboratory Director, Natera



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

Patient Information

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

RECOMMENDATIONS

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

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DEAFNESS, AUTOSOMAL RECESSIVE 77**Understanding Your Horizon Carrier Screen Results****What is Deafness, Autosomal Recessive 77?**

Deafness, Autosomal Recessive 77 is an inherited disorder that affects hearing. Affected individuals usually develop hearing loss beginning in childhood. The hearing loss worsens with age. This condition does not cause other health problems. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Deafness, Autosomal Recessive 77?

Deafness, Autosomal Recessive 77 is caused by a change, or mutation, in both copies of the LOXHD1 gene pair. These mutations cause the genes to not work properly or not work at all. Normal function of the LOXHD1 genes is important for hearing. When both copies of the LOXHD1 gene do not work correctly, progressive hearing loss occurs. Deafness, Autosomal Recessive 77 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 LOXHD1 gene to have a child with Deafness, Autosomal Recessive 77. People who are carriers for Deafness, Autosomal Recessive 77 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 Deafness, Autosomal Recessive 77, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their LOXHD1 gene mutations to the child, who will then have this condition. Individuals found to carry more than one mutation for Deafness, Autosomal Recessive 77 should discuss their risk for having an affected child, and any potential effects to their own hearing, with their health care provider. There are many other forms of inherited hearing loss, each caused by mutations in other genes. A person who is a carrier for Deafness, Autosomal Recessive 77 is not likely to be at increased risk for having a child with these other forms.

What can I do next?

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website (www.nsgc.org). Your siblings and other relatives are at increased risk to also have this mutation. You are encouraged to inform your family members of your test results as they may wish to consider being tested themselves. If you are pregnant, your partner can have carrier screening for Deafness, Autosomal Recessive 77 ordered by a health care professional. If your partner is not found to be a carrier for Deafness, Autosomal Recessive 77, your risk of having an affected child is greatly reduced. Couples at risk of having a baby with Deafness, Autosomal Recessive 77 can opt to have prenatal diagnosis done through chorionic villus sampling (CVS) or amniocentesis during pregnancy or can choose have the baby tested after birth. If you are not yet pregnant, your partner can have carrier screening for Deafness, Autosomal Recessive 77 ordered by a health care professional. If your partner is found to be a carrier for Deafness, Autosomal Recessive 77, you have several reproductive options to consider:

- Natural pregnancy with or without prenatal diagnostic testing of the fetus or testing the baby after birth for Deafness, Autosomal Recessive 77
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for Deafness, Autosomal Recessive 77
- Adoption or use of a sperm or egg donor who is not a carrier for Deafness, Autosomal Recessive 77

What resources are available?

- Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/gene/LOXHD1>
- Prenatal diagnosis by CVS: <http://www.marchofdimes.org/chorionic-villus-sampling.aspx>
- Prenatal diagnosis by amniocentesis: <http://www.marchofdimes.org/amniocentesis.aspx>
- PGD with IVF: <http://www.natera.com/spectrum>

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HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED**Understanding Your Horizon Carrier Screen Results****What is Hermansky-Pudlak Syndrome, HPS1-Related?**

Hermansky-Pudlak Syndrome, HPS1-Related is an inherited disorder that causes albinism (decreased color, or pigment, in the skin, hair, and eyes), bleeding problems, and may cause pulmonary fibrosis (scarring in the lungs). The lack of pigment (albinism) causes abnormal eye movements (nystagmus), vision problems, and an increased risk for skin cancer. People with this condition also have problems with blood clotting, leading to bruising and easy bleeding. Pulmonary fibrosis occurs in some people and typically begins in around the age of 30; the lung scarring leads to breathing problems that often result in death within about ten years after symptoms begin. 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 Hermansky-Pudlak Syndrome, HPS1-Related?

Hermansky-Pudlak Syndrome, HPS1-Related is caused by gene changes, or mutations, in both copies of the HPS1 gene pair. The function of the HPS1 genes is to help make pigment in the skin, hair and eyes, as well as to help with blood clotting. When both copies of the HPS1 gene are not working, it causes the symptoms described above. Hermansky-Pudlak Syndrome, HPS1-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 HPS1 gene to have a child with Hermansky-Pudlak Syndrome, HPS1-Related. People who are carriers for Hermansky-Pudlak Syndrome, HPS1-Related are usually healthy and do not have symptoms of the condition nor do they have Hermansky-Pudlak Syndrome, HPS1-Related 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 Hermansky-Pudlak Syndrome, HPS1-Related there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their HPS1 gene mutations to the child, who will then have this condition. Individuals found to carry more than one mutation for Hermansky-Pudlak Syndrome, HPS1-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 a number of other forms of Hermansky-Pudlak Syndrome, each caused by mutations in different genes. A person who is a carrier for Hermansky-Pudlak Syndrome, HPS1-Related is not likely to be at significantly increased risk for having a child with these other forms.

What can I do next?

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website (www.nsgc.org). Your siblings and other relatives are at increased risk to also have this mutation. You are encouraged to inform your family members of your test results as they may wish to consider being tested themselves. If you are pregnant, your partner can have carrier screening for Hermansky-Pudlak Syndrome, HPS1-Related ordered by a health care professional. If your partner is not found to be a carrier for Hermansky-Pudlak Syndrome, HPS1-Related, your risk of having a child with this condition is greatly reduced. Couples at risk of having a baby with Hermansky-Pudlak Syndrome, HPS1-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 Hermansky-Pudlak Syndrome, HPS1-Related ordered by a health care professional. If your partner is found to be a carrier for Hermansky-Pudlak Syndrome, HPS1-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 Hermansky-Pudlak Syndrome, HPS1-Related
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for Hermansky-Pudlak Syndrome, HPS1-Related
- Adoption or use of a sperm or egg donor who is not a carrier for Hermansky-Pudlak Syndrome, HPS1-Related

What resources are available?

- Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/hermansky-pudlak-syndrome>
- Hermansky-Pudlak Syndrome Network, Inc: <https://www.hpsnetwork.org/en/about-the-network/our-mission>
- 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>

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LEBER CONGENITAL AMAUROSIS TYPE GUCY2D

Understanding Your Horizon Carrier Screen Results

What do my results mean?

Your result shows that you have a change, or variant in the GUCY2D gene. Most variants mean that you are a carrier of GUCY2D-related conditions, one of which is Leber congenital amaurosis, GUCY2D-related (or LCA1). 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. If you are a carrier, your children are not at high risk for GUCY2D-related conditions unless your partner or donor is also a carrier of these conditions. Further testing can be done to see if your partner or donor is a carrier.

Certain variants in GUCY2D mean that you could have cone-rod dystrophy 6 (CORD6). If you have CORD6, your children are at risk of having this condition, but you are not certain to have a child with CORD6.

Sometimes your lab report provides more information about the variant you have. It could tell you if you are a carrier of GUCY2D-related conditions or if you and your children are at risk for CORD6.

What are GUCY2D-related conditions?

All GUCY2D-related conditions affect the eyes and vision. There are several GUCY2D-related conditions.

- **Leber congenital amaurosis, GUCY2D-related (LCA1)** People with LCA1 usually have severe vision loss from birth. Some people with LCA1 have vision loss starting later in life and milder symptoms. Most people with LCA1 are very sensitive to light, and many have cloudy lenses (cataracts) and shaky eye movements. Less frequently, people with LCA1 can have poor night vision and problems with color vision. Vision loss for people with LCA1 can get worse over time.^{1,2,3,4}
- **Early-onset retinitis pigmentosa (EORP)** People with EORP have poor overall vision starting in early childhood. They often have poor night and color vision, and a loss of side (peripheral) vision.⁴
- **Congenital night blindness (CNB)** People with CNB have night blindness starting soon after birth or in childhood. They can also have problems with color vision.^{4,5}
- **Cone-rod dystrophy 6 (CORD6)** People with CORD6 have vision loss that gets worse over time. Many people with CORD6 start having symptoms in childhood, but the vision loss can start at any age. The first symptom of CORD6 is usually that vision becomes less sharp. People with CORD6 are sensitive to light and can have problems with color vision. People with CORD6 eventually lose their central vision, and some lose their peripheral vision as well. Over time, many people with CORD6 develop shaky eye movements and night blindness. Most people with CORD6 are legally blind by mid-adulthood.^{6,7,8}

Currently there is no cure for GUCY2D-related conditions, and treatment is based on symptoms.² Clinical trials involving potential new treatments for these conditions could be available (see clinicaltrials.gov).

What causes GUCY2D-related conditions?

LCA1, EORP, and CNB are caused by certain changes, or variants, in the GUCY2D 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 GUCY2D gene. Carriers of these GUCY2D-related conditions have one working copy and one nonworking copy of the gene. People with these GUCY2D-related conditions have no working copies of the gene. It is sometimes, but not always, possible to tell if a specific variant will cause LCA1, EORP, or CNB.

LCA1, EORP, and CNB are usually passed down, or inherited, from both genetic parents. We inherit one copy of the GUCY2D 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 nonworking genes and having GUCY2D-related conditions. Each child also has a 1 in 2 (50%) chance of being a carrier of GUCY2D-related conditions and a 1 in 4 (25%) chance of inheriting two working copies of the gene. This type of inheritance is called autosomal recessive inheritance.

People with CORD6 have the condition because they have a specific kind of change in just one copy of the GUCY2D gene that causes it not to work properly. When one genetic parent has CORD6, each child has a 1 in 2 (50%) chance of getting the nonworking gene and having the condition. Each child also has a 1 in 2 (50%) chance of getting the working gene and not having the condition. This type of inheritance is called autosomal dominant inheritance.

Will my children have GUCY2D-related conditions?

If you are a carrier of GUCY2D-related conditions and your partner or donor is also a carrier of GUCY2D-related conditions, your children could have LCA1, EORP, or CNB. Each child you have together would have a 1 in 4 (25%) chance of having one of these conditions. Each child you have together would also have a 3 in 4 (75%) chance of **not** having one of these conditions. If your partner or donor has GUCY2D carrier screening and no variants are found, the chance that your children would have GUCY2D-related conditions is very low. No further testing would usually be needed for you, your partner or donor, or your children related to GUCY2D-related conditions.

If you have CORD6, each of your children has a 1 in 2 (50%) chance of having CORD6. Each of your children also has a 1 in 2 (50%) chance of **not** having this

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condition.

What can I do next?

Please speak to your healthcare provider about your result and whether it could impact your health.

If you are a GUCY2D-related conditions carrier and you want to know if your children are at risk for GUCY2D-related conditions, your partner or donor would need to have GUCY2D 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 GUCY2D-related conditions carrier, your children would be at risk for having GUCY2D-related conditions.

If you have **CORD6**, your children are at risk for having **CORD6**.

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 GUCY2D-related conditions. These tests both have a small risk of miscarriage. Babies can also be tested for GUCY2D-related conditions after birth instead.

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

- natural pregnancy with CVS or amniocentesis to test for GUCY2D-related conditions during pregnancy;
- natural pregnancy and testing the baby after birth for GUCY2D-related conditions;
- preimplantation genetic testing (PGT-M) with in vitro fertilization (IVF) to test embryos for GUCY2D-related conditions;
- adoption; or
- use of a sperm or egg donor who had no variants found in GUCY2D carrier screening.

Where can I find more information?

- The Foundation American Society of Retina Specialists [asrs.org/patients/retinal-diseases/37/leber-congenital-amaurosis-lca](https://www.asrs.org/patients/retinal-diseases/37/leber-congenital-amaurosis-lca)
- Foundation Fighting Blindness [fightingblindness.org](https://www.fightingblindness.org)
- CVS [marchofdimes.org/chorionic-villus-sampling](https://www.marchofdimes.org/chorionic-villus-sampling)
- Amniocentesis [marchofdimes.org/pregnancy/amniocentesis](https://www.marchofdimes.org/pregnancy/amniocentesis)

What does this mean for my family?

You likely got (inherited) this nonworking 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 GUCY2D carrier screening. If you have **CORD6**, your family members could also have the condition.

References

1. Online Mendelian Inheritance in Man, OMIM®. Johns Hopkins University, Baltimore, MD. LEBER CONGENITAL AMAUROSIS 1; LCA1. MIM Number: 204000. 12/17/2024. Available from: <https://omim.org/entry/204000>. Accessed December 2024.
2. Kumaran N et al. Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview. 2018 Oct 4 [Updated 2023 Mar 23]. In: Adam MP et al, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK531510/>. Accessed October 2024.
3. Huang C et al. Leber's Congenital Amaurosis: Current Concepts of Genotype-Phenotype Correlations. Genes (Basel). 2021 Aug 19;12(8):1261. doi: 10.3390/genes12081261. PMID: 34440435; PMCID: PMC8392113.
4. Rodilla C et al. Comprehensive Genotyping and Phenotyping Analysis of GUCY2D-Associated Rod- and Cone-Dominated Dystrophies. Am J Ophthalmol. 2023 Oct;254:87-103. doi: 10.1016/j.ajo.2023.05.015. Epub 2023 Jun 15. PMID: 37327959.
5. Online Mendelian Inheritance in Man, OMIM®. Johns Hopkins University, Baltimore, MD. NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE11; CSNB11. MIM Number: 618555. 11/7/2022. Available from: <https://omim.org/entry/618555/>. Accessed December 2024.
6. MedlinePlus Genetics [Internet]. Bethesda (MD): National Library of Medicine (US). Cone-rod dystrophy; [updated 2018 Mar 1; cited 2024 Dec 13]; [about 3 p.]. Available from: <https://medlineplus.gov/genetics/condition/cone-rod-dystrophy/#causes>.
7. Neubauer J et al. GUCY2D-Related Retinal Dystrophy with Autosomal Dominant Inheritance-A Multicenter Case Series and Review of Reported Data. Genes (Basel). 2022 Feb 8;13(2):313. doi: 10.3390/genes13020313. PMID: 35205358; PMCID: PMC8872159.
8. Online Mendelian Inheritance in Man, OMIM®. Johns Hopkins University, Baltimore, MD. CONE-ROD DYSTROPHY 6; CORD6. MIM Number: 601777. 4/3/2024. Available from: <https://omim.org/entry/601777>. Accessed December 2024.

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NON-SYNDROMIC HEARING LOSS, GJB2-RELATED**Understanding Your Horizon Carrier Screen Results****What is Non-Syndromic Hearing Loss, GJB2-Related?**

Non-Syndromic Hearing Loss, GJB2-Related (also called DFNB1) is an inherited disorder that causes early-onset hearing loss. "Non-syndromic" means that no other parts of the body are affected, making hearing loss the only symptom of this condition. In Non-Syndromic Hearing Loss, GJB2-Related, hearing loss is typically present at birth (congenital). However, some children have normal hearing at birth and develop hearing loss during childhood. The severity varies from mild to profound sensorineural hearing loss. The treatment for hearing loss includes hearing aids and, in some cases, cochlear implants. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov). Non-Syndromic Hearing Loss, GJB2-Related does not cause other health problems.

What causes Non-Syndromic Hearing Loss, GJB2-Related?

Non-Syndromic Hearing Loss, GJB2-Related is caused by a gene change, or mutation, in both copies of the GJB2 gene pair (also known as DFNB1). These mutations cause the genes to not work properly or not work at all. The function of the GJB2 genes is to make a protein that is important for hearing. When both copies of the GJB2 gene do not work correctly, it leads to Non-Syndromic Hearing Loss, GJB2- Related. Non-Syndromic Hearing Loss, GJB2-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 GJB2 gene to have a child with Non-Syndromic Hearing Loss, GJB2-Related. People who are carriers for Non-Syndromic Hearing Loss, GJB2-Related are usually healthy and usually do not have Non-Syndromic Hearing Loss 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 Non-Syndromic Hearing Loss, GJB2- Related, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their GJB2 gene mutations to the child, who will then have Non-Syndromic Hearing Loss, GJB2-Related. Very rarely, carriers of a single GJB2 mutation will have inherited hearing loss with or without other symptoms. These individuals usually have one parent who is also affected. This type of inheritance, where having only one mutation causes symptoms, is called autosomal dominant. When a person with autosomal dominant hearing loss has a child, there is a 50%, or 1 in 2, chance with each pregnancy of having a child who will also develop this type of hearing loss. It is sometimes, but not always, possible to determine whether a specific mutation in the GJB2 gene will cause autosomal recessive Non-Syndromic Hearing Loss or an autosomal dominant type of hearing loss. Individuals found to carry more than one mutation for Non-Syndromic Hearing Loss, GJB2-Related should discuss their risk for having an affected child and any potential effects to their own hearing 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). You may wish to share your carrier screening results with your health care providers, especially if you have a family history of hearing loss or have concerns about your own hearing. 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 Non-Syndromic Hearing Loss, GJB2- Related ordered by a health care professional. If your partner is not found to be a carrier for Non-Syndromic Hearing Loss, GJB2-Related, your risk of having a child with Non-Syndromic Hearing Loss, GJB2-Related is greatly reduced. Couples at risk of having a baby with Non-Syndromic Hearing Loss, GJB2-Related can opt to have prenatal diagnosis done through chorionic villus sampling (CVS) or amniocentesis during pregnancy or can choose to test the baby after birth for this condition. If you are not yet pregnant, your partner can have carrier screening for Non-Syndromic Hearing Loss, GJB2-Related ordered by a health care professional. If your partner is found to be a carrier for Non-Syndromic Hearing Loss, GJB2-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 Non-Syndromic Hearing Loss, GJB2- Related
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for Non-Syndromic Hearing Loss, GJB2-Related
- Adoption or use of a sperm or egg donor who is not a carrier for Non-Syndromic Hearing Loss, GJB2- Related

What resources are available?

- Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/nonsyndromic-hearing-loss>
- 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

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Case File ID: [REDACTED]

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THYROID DYSHORMONOGENESIS 6

Understanding Your Horizon Carrier Screen Results

What does being a carrier mean?

Your results show that you are a carrier of thyroid dysphormonogenesis 6 (TDH6). Most people with a variant in this gene are carriers of TDH6, but do not have the condition. Some people with a variant in this gene have symptoms of TDH6 as babies that go away as they age.

Your children are at risk for TDH6 or for short-term symptoms of this condition, but you are not certain to have a child with this condition. Further testing can be done to see if your partner or donor is a carrier.

What is thyroid dysphormonogenesis 6 (TDH6)?

TDH6 causes the body to not make enough thyroid hormones, resulting in congenital hypothyroidism (CH).^{1,2} Some people with CH have no symptoms. Other people with CH can be less active, sleep more than normal, and have feeding problems or constipation. People with CH that is not treated can also have slow growth and intellectual disability.² With early treatment, people with TDH6 usually have normal development.³ Newborn screening can detect over 90% of babies with CH.⁴

Carriers of TDH6 can have mild hypothyroidism as babies. Thyroid hormone levels can be lower than average at birth and increase with age.^{1,2}

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

What causes thyroid dysphormonogenesis 6 (TDH6)?

TDH6 is caused by changes, or variants, in the DUOX2 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 DUOX2 gene. Carriers of TDH6 have one working copy and one non-working copy of the gene. Some carriers have low levels of thyroid hormones as babies, but have normal thyroid function as they get older. People with TDH6 have no working copies of the gene.

TDH6 is usually passed down, or inherited, from both genetic parents. We inherit one copy of the DUOX2 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 TDH6. Each child also has a 1 in 2 (50%) chance of being a carrier of TDH6 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 thyroid dysphormonogenesis 6 (TDH6)?

If your partner or donor also has a non-working copy of the DUOX2 gene, your children could have TDH6. Each child you have together would have a 1 in 4 (25%) chance of having TDH6. Each child you have together would also have a 1 in 4 (25%) chance of **not** having any variants in the DUOX2 gene. Each child would have a 1 in 2 (50%) chance of being a carrier and could have symptoms of the condition as a baby.

If your partner or donor has DUOX2 carrier screening and no variants are found, the chance that your children would have two TDH6 variants is very low. In this situation, each child you have together would have a 1 in 2 (50%) chance of being a carrier and could have symptoms of TDH6 as a baby.

What can I do next?

If you want to know if your children are at risk for TDH6, your partner or donor would need to have DUOX2 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 TDH6 carrier, your children would be at risk for having TDH6. Your children are also at risk of being carriers who have low levels of thyroid hormones as babies.

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 TDH6. These tests both have a small risk of miscarriage. Babies can also be tested for TDH6 after birth instead.

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

- natural pregnancy with CVS or amniocentesis to test for TDH6 during pregnancy;
- natural pregnancy and testing the baby after birth for TDH6;
- preimplantation genetic testing (PGT-M) with in vitro fertilization (IVF) to test embryos for TDH6;
- adoption; or
- use of a sperm or egg donor who had no variants found in DUOX2 carrier screening.

Where can I find more information?

- Pediatric Endocrine Society pedsendo.org/patient-resource/congenital-hypothyroidism

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- American Thyroid Association thyroid.org/professionals
- 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 TDH6.

References

1. Moreno JC et al. Inactivating mutations in the gene for thyroid oxidase 2 (THOX2) and congenital hypothyroidism. *New Eng. J. Med.* 347: 95-102, 2002.
2. Vigone MC et al. Persistent mild hypothyroidism associated with novel sequence variants of the DUOX2 gene in two siblings. *Hum. Mutat.* 26: 395, 2005.
3. MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US). Congenital hypothyroidism; [updated 2015 Sep 1; cited 2024 March 3]. Available from: <https://medlineplus.gov/genetics/condition/congenital-hypothyroidism/>.
4. Büyükgebiz A. Newborn screening for congenital hypothyroidism. *J Clin Res Pediatr Endocrinol.* 2013;5 Suppl 1(Suppl 1):8-12. doi: [10.4274/jcrpe.845](https://doi.org/10.4274/jcrpe.845). Epub 2012 Nov 15. PMID: 23154158; PMCID: PMC3608007.

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**VARIANT DETAILS****DUOX2, c.514-1G>A, likely pathogenic**

- The c.514-1G>A variant in the DUOX2 gene has been observed at a frequency of 0.0008% in the gnomAD v2.1.1 dataset.
- This canonical splicing variant is predicted to alter the reading frame and cause nonsense-mediated decay (NMD) in a gene where loss-of-function is a known mechanism of disease.
- This variant has not been described in ClinVar.

GJB2, c.109G>A (p.V37I), pathogenic

- The c.109G>A (p.V37I) variant in the GJB2 gene has been observed at a frequency of 0.7556% in the gnomAD v2.1.1 dataset.
- This variant has been reported in a homozygous state or in conjunction with another variant in individuals with mild late-onset hearing loss with reduced penetrance (PMID: 17036313, 17935238).
- This variant has been reported in ClinVar [ID: 17023].

GUCY2D, c.2927G>T (p.R976L), likely pathogenic

- The c.2927G>T (p.R976L) variant in the GUCY2D gene has been observed at a frequency of 0.0004% in the gnomAD v2.1.1 dataset.
- This variant has been reported in a homozygous state or in conjunction with another variant in individual(s) with GUCY2D-related Leber congenital amaurosis 2 (PMID: 10951519, 17525851).
- Functional studies demonstrated this variant abolishes GUCY2D catalytic activity (PMID: 11328726).
- This variant has been described in ClinVar [ID: 98581].

HPS1, c.9C>A (p.C3*), pathogenic

- The c.9C>A (p.C3*) variant in the HPS1 gene has not been observed in the gnomAD v2.1.1 dataset.
- This variant has been reported in a homozygous state or in conjunction with another variant in individual(s) with Hermansky-Pudlak syndrome, type 1 (PMID: 33124154).
- This premature termination variant is predicted to cause nonsense-mediated decay (NMD) in a gene where loss-of-function is a known mechanism of disease.
- This variant has been reported in ClinVar [ID: 1324541].

LOXHD1, c.3281A>G (p.D1094G), likely pathogenic

- The c.3281A>G (p.D1094G) variant in the LOXHD1 gene has been observed at a frequency of 0.0052% in the gnomAD v2.1.1 dataset.
- This variant has been reported in a homozygous state or in conjunction with another variant in individual(s) with nonsyndromic hearing loss and deafness (DFNB) 77 (PMID: 31547530).
- This variant has been reported in ClinVar [ID: 891722].

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DISEASES SCREENED

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

Autosomal Recessive

- 1**
17-BETA HYDROXYSTEROID DEHYDROGENASE 3 DEFICIENCY (*HSD17B3*) **negative**
- 3**
3-BETA-HYDROXYSTEROID DEHYDROGENASE TYPE II DEFICIENCY (*HSD3B2*) **negative**
3-HYDROXY-3-METHYLGLUTARYL-COENZYME A LYASE DEFICIENCY (*HMGCL*) **negative**
3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (*HADH*) **negative**
3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY (*MCCC2*) **negative**
3-PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY (*PHGDH*) **negative**
- 5**
5-ALPHA-REDUCTASE DEFICIENCY (*SRD5A2*) **negative**
- 6**
6-PYRUVOYL-TETRAHYDROPTERIN SYNTHASE (*PTPS*) DEFICIENCY (*PTS*) **negative**
- A**
ABCA4-RELATED CONDITIONS (*ABCA4*) **negative**
ABETALIPOPROTEINEMIA (*MTPP*) **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-GOUTIÈRES SYNDROME, RNASEH2A-RELATED (*RNASEH2A*) **negative**
AICARDI-GOUTIÈRES SYNDROME, RNASEH2B-RELATED (*RNASEH2B*) **negative**
AICARDI-GOUTIÈRES SYNDROME, RNASEH2C-RELATED (*RNASEH2C*) **negative**
AICARDI-GOUTIÈRES SYNDROME, TREX1-RELATED (*TREX1*) **negative**
ALPHA-MANNOSIDOSIS (*MAN2B1*) **negative**
ALPHA-THALASSEMIA (*HBA1/HBA2*) **negative**
ALPORT SYNDROME, COL4A3-RELATED (*COL4A3*) **negative**
ALPORT SYNDROME, COL4A4-RELATED (*COL4A4*) **negative**
ALSTROM SYNDROME (*ALMS1*) **negative**
AMISH INFANTILE EPILEPSY SYNDROME (*ST3GAL5*) **negative**
ANDERMANN SYNDROME (*SLC12A6*) **negative**
ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (*GATM*) **negative**
ARGININEMIA (*ARG1*) **negative**
ARGININOSUCCINATE LYASE DEFICIENCY (*ASL*) **negative**
AROMATASE DEFICIENCY (*CYP19A1*) **negative**
ASPARAGINE SYNTHETASE DEFICIENCY (*ASNS*) **negative**
ASPARTYLGLYCOSAMINURIA (AGA) **negative**
ATAXIA WITH VITAMIN E DEFICIENCY (*TTPA*) **negative**
ATAXIA-TELANGIECTASIA (*ATM*) **negative**
ATAXIA-TELANGIECTASIA-LIKE DISORDER 1 (*MRE11*) **negative**
ATRANSFERRINEMIA (*TF*) **negative**
AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (*SLC35A3*) **negative**
AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (*AIRE*) **negative**
AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (*ARCI*), SLC27A4-RELATED (*SLC27A4*) **negative**
AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (*SACS*) **negative**
- B**
BARDET-BIEDL SYNDROME, ARL6-RELATED (*ARL6*) **negative**
- BARDET-BIEDL SYNDROME, BBS10-RELATED (*BBS10*) **negative**
BARDET-BIEDL SYNDROME, BBS12-RELATED (*BBS12*) **negative**
BARDET-BIEDL SYNDROME, BBS1-RELATED (*BBS1*) **negative**
BARDET-BIEDL SYNDROME, BBS2-RELATED (*BBS2*) **negative**
BARDET-BIEDL SYNDROME, BBS4-RELATED (*BBS4*) **negative**
BARDET-BIEDL SYNDROME, BBS5-RELATED (*BBS5*) **negative**
BARDET-BIEDL SYNDROME, BBS7-RELATED (*BBS7*) **negative**
BARDET-BIEDL SYNDROME, BBS9-RELATED (*BBS9*) **negative**
BARDET-BIEDL SYNDROME, TTC8-RELATED (*TTC8*) **negative**
BARE LYMPHOCYTE SYNDROME, CIITA-RELATED (*CIITA*) **negative**
BARTTER SYNDROME, BSND-RELATED (*BSND*) **negative**
BARTTER SYNDROME, KCNJ1-RELATED (*KCNJ1*) **negative**
BARTTER SYNDROME, SLC12A1-RELATED (*SLC12A1*) **negative**
BATTEN DISEASE, CLN3-RELATED (*CLN3*) **negative**
BETA-HEMOGLOBINOPATHIES (*HBB*) **negative**
BETA-KETOTHIOLASE DEFICIENCY (*ACAT1*) **negative**
BETA-MANNOSIDOSIS (*MANBA*) **negative**
BETA-UREIDOPROPIONASE DEFICIENCY (*UPB1*) **negative**
BILATERAL FRONTOPARIETAL POLYMICROGYRIA (*GPR56*) **negative**
BIOTINIDASE DEFICIENCY (*BTD*) **negative**
BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBGD) (*SLC19A3*) **negative**
BLOOM SYNDROME (*BLM*) **negative**
BRITTLE CORNEA SYNDROME 1 (*ZNF469*) **negative**
BRITTLE CORNEA SYNDROME 2 (*PRDM5*) **negative**
- C**
CANAVAN DISEASE (*ASPA*) **negative**
CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (*CPS1*) **negative**
CARNITINE DEFICIENCY (*SLC22A5*) **negative**
CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY (*CPT1A*) **negative**
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (*CPT2*) **negative**
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (*SLC25A20*) **negative**
CARPENTER SYNDROME (*RAB23*) **negative**
CARTILAGE-HAIR HYPOPLASIA (*RMRP*) **negative**
CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (*CASQ2*) **negative**
CD59-MEDIATED HEMOLYTIC ANEMIA (*CD59*) **negative**
CEP152-RELATED MICROCEPHALY (*CEP152*) **negative**
CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA (CEDNIK) SYNDROME (*SNAP29*) **negative**
CEREBROTENDINOUS XANTHOMATOSIS (*CYP27A1*) **negative**
CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE C (*PLEKHG5*) **negative**
CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (*NDRG1*) **negative**
CHEDIAK-HIGASHI SYNDROME (*LYST*) **negative**
CHOREOACANTHOCYTOSIS (*VPS13A*) **negative**
CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (*CYBA*) **negative**
CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (*NCF2*) **negative**
CILIOPATHIES, RPGRIP1L-RELATED (*RPGRIP1L*) **negative**
CITRIN DEFICIENCY (*SLC25A13*) **negative**
CITRULLINEMIA, TYPE 1 (*ASS1*) **negative**
CLN10 DISEASE (*CTS1D*) **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 (*TFSM*) **negative**

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C

COMBINED PITUITARY HORMONE DEFICIENCY 1 (*POU1F1*) **negative**
 COMBINED PITUITARY HORMONE DEFICIENCY-2 (*PROP1*) **negative**
 CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (*CYP11B1*) **negative**
 CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (*CYP17A1*) **negative**
 CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (*CYP21A2*) **negative**
 CONGENITAL ADRENAL INSUFFICIENCY, *CYP11A1*-RELATED (*CYP11A1*) **negative**
 CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (*MPL*) **negative**
 CONGENITAL CHRONIC DIARRHEA (*DGAT1*) **negative**
 CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, *ALG1*-RELATED (*ALG1*) **negative**
 CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, *PMM2*-Related (*PMM2*) **negative**
 CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (*MPL*) **negative**
 CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (*ALG6*) **negative**
 CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (*SEC23B*) **negative**
 CONGENITAL FINNISH NEPHROSIS (*NPHS1*) **negative**
 CONGENITAL HYDROCEPHALUS 1 (*CCDC88C*) **negative**
 CONGENITAL HYPERINSULINISM, *KCNJ11*-Related (*KCNJ11*) **negative**
 CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (*CIPA*) (*NTRK1*) **negative**
 CONGENITAL MYASTHENIC SYNDROME, *CHAT*-RELATED (*CHAT*) **negative**
 CONGENITAL MYASTHENIC SYNDROME, *CHRNE*-RELATED (*CHRNE*) **negative**
 CONGENITAL MYASTHENIC SYNDROME, *COLQ*-RELATED (*COLQ*) **negative**
 CONGENITAL MYASTHENIC SYNDROME, *DOK7*-RELATED (*DOK7*) **negative**
 CONGENITAL MYASTHENIC SYNDROME, *RAPSN*-RELATED (*RAPSN*) **negative**
 CONGENITAL NEPHROTIC SYNDROME, *PLCE1*-RELATED (*PLCE1*) **negative**
 CONGENITAL NEUTROPENIA, *G6PC3*-RELATED (*G6PC3*) **negative**
 CONGENITAL NEUTROPENIA, *HAX1*-RELATED (*HAX1*) **negative**
 CONGENITAL NEUTROPENIA, *VPS45*-RELATED (*VPS45*) **negative**
 CONGENITAL SECRETORY CHLORIDE DIARRHEA 1 (*SLC26A3*) **negative**
 CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS (*SLC4A11*) **negative**
 CORTICOSTERONE METHYLOXIDASE DEFICIENCY (*CYP11B2*) **negative**
 COSTEFF SYNDROME (3-METHYLG LUTACONIC ACIDURIA, TYPE 3) (*OPA3*) **negative**
CRB1-RELATED RETINAL DYSTROPHIES (*CRB1*) **negative**
 CYSTIC FIBROSIS (*CFTR*) **negative**
 CYSTINOSIS (*CTNS*) **negative**
 CYTOCHROME C OXIDASE DEFICIENCY, *PET100*-RELATED (*PET100*) **negative**
 CYTOCHROME P450 OXIDOREDUCTASE DEFICIENCY (*POR*) **negative**

D

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

E

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

F

FACTOR XI DEFICIENCY (*F11*) **negative**

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

G

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

H

HARLEQUIN ICHTHYOSIS (*ABCA12*) **negative**
 HEME OXYGENASE 1 DEFICIENCY (*HMOX1*) **negative**
 HEMOCHROMATOSIS TYPE 2A (*HFE2*) **negative**
 HEMOCHROMATOSIS, TYPE 3, *TFR2*-Related (*TFR2*) **negative**
 HEPATOCEREBRAL MITOCHONDRIAL DNA DEPLETION SYNDROME, *MPV17*-RELATED (*MPV17*) **negative**

Patient Information

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Test Information

Ordering Physician: [REDACTED]



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H

HEREDITARY FRUCTOSE INTOLERANCE (ALDOB) **negative**
 HEREDITARY HEMOCHROMATOSIS TYPE 2B (HAMP) **negative**
 HEREDITARY SPASTIC PARAPLEGIA, TYPE 49 (TECPR2) **negative**
 HEREDITARY SPASTIC PARAPLEGIA, CYP7B1-RELATED (CYP7B1) **negative**
 HERMANSKY-PUDLAK SYNDROME, AP3B1-RELATED (AP3B1) **negative**
 HERMANSKY-PUDLAK SYNDROME, BLOC153-RELATED (BLOC153) **negative**
 HERMANSKY-PUDLAK SYNDROME, BLOC156-RELATED (BLOC156) **negative**
 HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED (HPS1) **see first page**
 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 (HMLS1) **negative**
 HYPER-IGM IMMUNODEFICIENCY (CD40) **negative**
 HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA (HHH SYNDROME) (SLC25A15) **negative**
 HYPERPHOSPHATEMIC FAMILIAL TUMORAL CALCINOSIS, GALNT3-RELATED (GALNT3) **negative**
 HYPOMYELINATING LEUKODYSTROPHY 12 (VPS11) **negative**
 HYPOPHOSPHATASIA, ALPL-RELATED (ALPL) **negative**

I

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

J

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

KKRABBE 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) **see first page**
 LEBER CONGENITAL AMAUROSIS TYPE TULP1 (TULP1) **negative**
 LEBER CONGENITAL AMAUROSIS, IQCB1-RELATED/SENIOR-LOKEN SYNDROME 5 (IQCB1) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE CEP290 (CEP290) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE LCA5 (LCA5) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE RDH12 (RDH12) **negative**
 LEIGH SYNDROME, FRENCH-CANADIAN TYPE (LRPPRC) **negative**
 LETHAL CONGENITAL CONTRACTURE SYNDROME 1 (GLE1) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER (EIF2B5) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B1-RELATED (EIF2B1) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B2-RELATED (EIF2B2) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B3-RELATED (EIF2B3) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B4-RELATED (EIF2B4) **negative**
 LIG4 SYNDROME (LIG4) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 8 (TRIM32) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2A (CAPN3) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2B (DYSF) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2C (SGCG) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2D (SGCA) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2E (SGCB) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2F (SGCD) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2I (FKRP) **negative**
 LIPOAMIDE DEHYDROGENASE DEFICIENCY (DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY) (DLD) **negative**
 LIPOID ADRENAL HYPERPLASIA (STAR) **negative**
 LIPOPROTEIN LIPASE DEFICIENCY (LPL) **negative**
 LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (HADHA) **negative**
 LRAT-RELATED CONDITIONS (LRAT) **negative**
 LUNG DISEASE, IMMUNODEFICIENCY, AND CHROMOSOME BREAKAGE SYNDROME (LICS) (NSMCE3) **negative**
 LYSINURIC PROTEIN INTOLERANCE (SLC7A7) **negative**

M

MALONYL-COA DECARBOXYLASE DEFICIENCY (MLYCD) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 1A (BCKDHA) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 1B (BCKDHB) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 2 (DBT) **negative**
 MCKUSICK-KAUFMAN SYNDROME (MKKS) **negative**
 MECKEL SYNDROME 7/NEPHRONOPHTHISIS 3 (NPHP3) **negative**
 MECKEL-GRUBER SYNDROME, TYPE 1 (MKS1) **negative**
 MECR-RELATED NEUROLOGIC DISORDER (MECR) **negative**
 MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (ACADM) **negative**
 MEDNIK SYNDROME (AP1S1) **negative**
 MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS (MLC1) **negative**
 MEROSIN-DEFICIENT MUSCULAR DYSTROPHY (LAMA2) **negative**
 METABOLIC ENCEPHALOPATHY AND ARRHYTHMIAS, TANGO2-RELATED (TANGO2) **negative**
 METACHROMATIC LEUKODYSTROPHY, ARSA-RELATED (ARSA) **negative**
 METACHROMATIC LEUKODYSTROPHY, PSAP-RELATED (PSAP) **negative**
 METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CBLF (LMBRD1) **negative**
 METHYLMALONIC ACIDEMIA, MCEE-RELATED (MCEE) **negative**
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CBLF (MMACHC) **negative**
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CblD (MMADHC) **negative**

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M

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 I DEFICIENCY, ACAD9-RELATED (ACAD9) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NDUFAF5-RELATED (NDUFAF5) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NDUFS6-RELATED (NDUFS6) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 1 (NDUFS4) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 10 (NDUFAF2) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 17 (NDUFAF6) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 19 (FOXRED1) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 3 (NDUFS7) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 4 (NDUFV1) **negative**
 MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 2, SCO2-RELATED (SCO2) **negative**
 MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 6 (COX15) **negative**
 MITOCHONDRIAL DNA DEPLETION SYNDROME 2 (TK2) **negative**
 MITOCHONDRIAL DNA DEPLETION SYNDROME 3 (DGUOK) **negative**
 MITOCHONDRIAL MYOPATHY AND SIDEROBLASTIC ANEMIA (MLASA1) (PUS1) **negative**
 MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (HADHB) **negative**
 MOLYBDENUM COFACTOR DEFICIENCY TYPE B (MOCS2) **negative**
 MOLYBDENUM COFACTOR DEFICIENCY, TYPE A (MOCS1) **negative**
 MUCOLIPIDOSIS II/III A (GNPTAB) **negative**
 MUCOLIPIDOSIS III GAMMA (GNPTG) **negative**
 MUCOLIPIDOSIS, TYPE IV (MCOLN1) **negative**
 MUCOPOLYSACCHARIDOSIS, TYPE I (HURLER SYNDROME) (IDUA) **negative**
 MUCOPOLYSACCHARIDOSIS, TYPE III A (SANFILIPPO A) (SGSH) **negative**
 MUCOPOLYSACCHARIDOSIS, TYPE III B (SANFILIPPO B) (NAGLU) **negative**
 MUCOPOLYSACCHARIDOSIS, TYPE III C (SANFILIPPO C) (HGSNAT) **negative**
 MUCOPOLYSACCHARIDOSIS, TYPE III D (SANFILIPPO D) (GNS) **negative**
 MUCOPOLYSACCHARIDOSIS, TYPE IV A (MORQUIO SYNDROME) (GALNS) **negative**
 MUCOPOLYSACCHARIDOSIS, TYPE IV B/GM1 GANGLIOSIDOSIS (GLB1) **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) **see first page**
 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, PJKV-RELATED (PJKV) **negative**
 NONSYNDROMIC HEARING LOSS, SYNE4-RELATED (SYNE4) **negative**
 NONSYNDROMIC HEARING LOSS, TMC1-RELATED (TMC1) **negative**
 NONSYNDROMIC HEARING LOSS, TMPRSS3-RELATED (TMPRSS3) **negative**
 NONSYNDROMIC INTELLECTUAL DISABILITY (CC2D1A) **negative**
 NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (SAMD9) **negative**

O

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

P

PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION (PANK2) **negative**
 PAPILLON LEFÈVRE SYNDROME (CTSC) **negative**
 PARKINSON DISEASE 15 (FBXO7) **negative**
 PENDRED SYNDROME (SLC26A4) **negative**
 PERLMAN SYNDROME (DIS3L2) **negative**
 PGM3-CONGENITAL DISORDER OF GLYCOSYLATION (PGM3) **negative**
 PHENYLKETONURIA (PAH) **negative**
 PIGN-CONGENITAL DISORDER OF GLYCOSYLATION (PIGN) **negative**
 PITUITARY HORMONE DEFICIENCY, COMBINED 3 (LHX3) **negative**
 POLG-RELATED DISORDERS (POLG) **negative**
 POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE (PKHD1) **negative**
 PONTOCEREBELLAR HYPOPLASIA, EXOSC3-RELATED (EXOSC3) **negative**
 PONTOCEREBELLAR HYPOPLASIA, RARS2-RELATED (RARS2) **negative**
 PONTOCEREBELLAR HYPOPLASIA, TSEN2-RELATED (TSEN2) **negative**
 PONTOCEREBELLAR HYPOPLASIA, TSEN54-RELATED (TSEN54) **negative**
 PONTOCEREBELLAR HYPOPLASIA, TYPE 1A (VRK1) **negative**
 PONTOCEREBELLAR HYPOPLASIA, TYPE 2D (SEPSECS) **negative**
 PONTOCEREBELLAR HYPOPLASIA, VPS53-RELATED (VPS53) **negative**
 PRIMARY CILIARY DYSKINESIA, CCDC103-RELATED (CCDC103) **negative**
 PRIMARY CILIARY DYSKINESIA, CCDC39-RELATED (CCDC39) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAH11-RELATED (DNAH11) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAH5-RELATED (DNAH5) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAI1-RELATED (DNAI1) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAI2-RELATED (DNAI2) **negative**
 PRIMARY CONGENITAL GLAUCOMA/PETERS ANOMALY (CYP1B1) **negative**
 PRIMARY HYPEROXALURIA, TYPE 1 (AGXT) **negative**
 PRIMARY HYPEROXALURIA, TYPE 2 (GRHPR) **negative**
 PRIMARY HYPEROXALURIA, TYPE 3 (HOGA1) **negative**
 PRIMARY MICROCEPHALY 1, AUTOSOMAL RECESSIVE (MCPH1) **negative**
 PROGRESSIVE EARLY-ONSET ENCEPHALOPATHY WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM (TBCD) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, ABCB4-RELATED (ABCB4) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 1 (PFIC1) (ATP8B1) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 2 (ABCB11) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 4 (PFIC4) (TJP2) **negative**
 PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA (CCN6) **negative**
 PROLIDASE DEFICIENCY (PEPD) **negative**
 PROPIONIC ACIDEMIA, PCCA-RELATED (PCCA) **negative**
 PROPIONIC ACIDEMIA, PCCB-RELATED (PCCB) **negative**
 PSEUDOXANTHOMA ELASTICUM (ABCC6) **negative**

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Test Information

Ordering Physician: [REDACTED]



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P

PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (PCD) DEFICIENCY (*PCBD1*) **negative**
 PYCNODYSTOSIS (*CTSK*) **negative**
 PYRIDOXAL 5'-PHOSPHATE-DEPENDENT EPILEPSY (*PNPO*) **negative**
 PYRIDOXINE-DEPENDENT EPILEPSY (*ALDH7A1*) **negative**
 PYRUVATE CARBOXYLASE DEFICIENCY (*PC*) **negative**
 PYRUVATE DEHYDROGENASE DEFICIENCY, PDHB-RELATED (*PDHB*) **negative**

R

REFSUM DISEASE, PHYH-RELATED (*PHYH*) **negative**
 RENAL TUBULAR ACIDOSIS AND DEAFNESS, ATP6V1B1-RELATED (*ATP6V1B1*) **negative**
 RENAL TUBULAR ACIDOSIS, PROXIMAL, WITH OCULAR ABNORMALITIES AND MENTAL
 RETARDATION (*SLC4A4*) **negative**
 RETINITIS PIGMENTOSA 25 (*EYS*) **negative**
 RETINITIS PIGMENTOSA 26 (*CERKL*) **negative**
 RETINITIS PIGMENTOSA 28 (*FAM161A*) **negative**
 RETINITIS PIGMENTOSA 36 (*PRCD*) **negative**
 RETINITIS PIGMENTOSA 59 (*DHDDS*) **negative**
 RETINITIS PIGMENTOSA 62 (*MAK*) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1 (*PEX7*) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2 (*GNPAT*) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3 (*AGPS*) **negative**
 RLB1-RELATED RETINOPATHY (*RLB1*) **negative**
 ROBERTS SYNDROME (*ESCO2*) **negative**
 RYR1-RELATED CONDITIONS (*RYR1*) **negative**

S

SALLA DISEASE (*SLC17A5*) **negative**
 SANDHOFF DISEASE (*HEXB*) **negative**
 SCHIMKE IMMUNOSKELETAL DYSPLASIA (*SMARCA1*) **negative**
 SCHINDLER DISEASE (*NAGA*) **negative**
 SEGAWA SYNDROME, TH-RELATED (*TH*) **negative**
 SENIOR-LOKEN SYNDROME 4/NEPHRONOPHTHISIS 4 (*NPHP4*) **negative**
 SEPIAPTERIN REDUCTASE DEFICIENCY (*SPR*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3D-RELATED (*CD3D*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3E-RELATED (*CD3E*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), FOXP1-RELATED (*FOXP1*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), IKBKB-RELATED (*IKBKB*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), IL7R-RELATED (*IL7R*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), JAK3-RELATED (*JAK3*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), PTPRC-RELATED (*PTPRC*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), RAG1-RELATED (*RAG1*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY, ADA-Related (*ADA*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY, TYPE ATHABASKAN (*DCLRE1C*) **negative**
 SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY
 (*DYNC2H1*) **negative**
 SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED (*SBDS*) **negative**
 SIALIDOSIS (*NEU1*) **negative**
 SJÖGREN-LARSSON SYNDROME (*ALDH3A2*) **negative**
 SMITH-LEMLI-OPITZ SYNDROME (*DHCR7*) **negative**
 SPASTIC PARAPLEGIA, TYPE 15 (*ZFYVE26*) **negative**
 SPASTIC TETRAPLEGIA, THIN CORPUS CALLOSUM, AND PROGRESSIVE MICROCEPHALY
 (*SPATCCM*) (*SLC1A4*) **negative**
 SPG11-RELATED CONDITIONS (*SPG11*) **negative**
 SPINAL MUSCULAR ATROPHY (*SMN1*) **negative** *SMN1: Two copies; g.27134T>G: absent; the
 absence of the g.27134T>G variant decreases the chance to be a silent (2+0) carrier.*
 SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS TYPE 1 (*IGHMBP2*) **negative**
 SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 10 (*ANO10*) **negative**
 SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12 (*WWOX*) **negative**
 SPONDYLOCOSTAL DYSOSTOSIS 1 (*DLL3*) **negative**
 SPONDYLOTORACIC 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*) **see first page**
 TRANSCOBALAMIN II DEFICIENCY (*TCN2*) **negative**
 TRICHOHEPATOENTERIC SYNDROME, SKIC2-RELATED (*SKIC2*) **negative**
 TRICHOHEPATOENTERIC SYNDROME, TTC37-RELATED (*TTC37*) **negative**
 TRICHOHODIODYSTROPHY 1/XERODERMA PIGMENTOSUM, GROUP D (*ERCC2*) **negative**
 TRIMETHYLAMINURIA (*FMO3*) **negative**
 TRIPLE A SYNDROME (*AAAS*) **negative**
 TSHR-RELATED CONDITIONS (*TSHR*) **negative**
 TYROSINEMIA TYPE III (*HPD*) **negative**
 TYROSINEMIA, TYPE 1 (*FAH*) **negative**
 TYROSINEMIA, TYPE 2 (*TAT*) **negative**

U

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

V

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

W

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

X

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

Z

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

Patient Information

Patient Name: DONOR 8208

Test Information

Ordering Physician: [REDACTED]



Date Of Birth: [REDACTED]

Clinic Information: Fairfax Cryobank

Case File ID: [REDACTED]

Report Date: 09/11/2025

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

Patient Information

Patient Name: DONOR 8208

Test Information

Ordering Physician: [REDACTED]



Date Of Birth: [REDACTED]

Clinic Information: Fairfax Cryobank

Case File ID: [REDACTED]

Report Date: 09/11/2025

Testing Methodology, Limitations, and Comments:**Next-generation sequencing (NGS)**

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

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

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

SPECIAL NOTES

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

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

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

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

For GJB2, CNV analysis of upstream deletions of GJB6-CRYL1 critical region is included.

For HBA1/HBA2, CNV analysis is offered to detect common deletions of -alpha3.7, -alpha4.2, --MED, --SEA, --FIL, --THAI, --alpha20.5, and/or HS-40. Sequencing and CNV analysis may have reduced sensitivity due to high sequence homology.

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

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

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

Patient Information

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Date Of Birth: [REDACTED]

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**Friedreich Ataxia (FXN)**

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

Friedreich Ataxia Repeat Categories

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

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.