Patient Information		Test Information	
Patient Name:		Ordering Physician:	Coralie Beauchamps
		Clinic Information:	Clinique Ovo
Date Of Birth:			
Gender:	Male		
Ethnicity:		Phone:	
Patient ID:	N/A	Report Date:	06/08/2025
Medical Record #:	N/A	Sample Collected:	05/27/2025
Collection Kit:		Sample Received:	05/28/2025
Accession ID:		Sample Type:	Blood
Case File ID:			

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:

SILENT CARRIER for Alpha-Thalassemia (aa/a-)

Positive for the pathogenic alpha 3.7 deletion of the HBA2 gene. Depending on the carrier status of the individual's partner, this couple may be at increased risk to have a child with Hemoglobin H Disease. Carrier screening for this individual's partner is suggested.

CARRIER for Congenital Dyserythropoietic Anemia Type 2

Positive for the pathogenic variant c.40C>T (p.R14W) in the SEC23B gene. If this individual's partner is a carrier for CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2, 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 Pseudocholinesterase Deficiency

Positive for the likely pathogenic variant c.635C>T (p.A212V) in the BCHE gene. Carriers of Pseudocholinesterase Deficiency may be at risk for adverse reactions to certain drugs and should inform their providers of their carrier status before having anesthesia (PMID 31082076). Comprehensive genetic counseling and additional medical workup as clinically indicated should be considered. If this individual's partner is a carrier for PSEUDOCHOLINESTERASE DEFICIENCY, 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 Wilson Disease

Positive for the likely pathogenic variant c.2479C>T (p.R827W) in the ATP7B gene. If this individual's partner is a carrier for WILSON DISEASE, 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 547 out of 551 diseases

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

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

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

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natera

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Patient Information	Test Information	
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ALPHA-THALASSEMIA SILENT CARRIER

Understanding Your Horizon Carrier Screen Results

What Is Alpha-Thalassemia?

Alpha-Thalassemia refers to a group of inherited blood disorders that reduce the amount of hemoglobin, the protein in red blood cells that carries oxygen to cells throughout the body. A person with one of the Alpha-Thalassemia diseases has lifelong anemia. Mild anemia can lead to tiredness, irritability, dizziness, lightheadedness and a rapid heartbeat. Severe anemia can be life threatening and may require routine blood transfusions. 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 these conditions may be available (see www.clinicaltrials.gov).

What causes Alpha-Thalassemia?

Hemoglobin is made of both alpha globin and beta globin proteins. There are four HBA genes (also called alpha globin genes) that are responsible for making alpha globin. Alpha-Thalassemia occurs when three or more of these four alpha globin genes is missing or changed. The exact type of Alpha- Thalassemia a person has depends on how many of the alpha globin genes are not working. Hemoglobin H Disease (a-/--): three missing or changed alpha globin genes. A person who has three missing or changed alpha globin genes has Hemoglobin H Disease. Hemoglobin H Disease can be mild or severe. People with severe disease may have chronic anemia, liver disease, and bone changes. Some people with Hemoglobin H Disease require frequent blood transfusions and other treatments. Alpha-Thalassemia Major, also known as Hemoglobin Bart's Disease (--/--): four missing or changed alpha globin genes. This results in severe fatal anemia. Affected babies develop symptoms before birth and without treatment typically do not survive the newborn period. Fetal blood transfusions during pregnancy may allow survival until after birth, at which time either lifelong transfusions or a stem cell transplantation will be necessary. Mothers who are pregnant with a fetus with Alpha-Thalassemia major can develop health problems during pregnancy. Alpha-Thalassemia is inherited in an autosomal recessive manner. Children typically inherit four copies of each alpha globin gene, two copies from the mother and two copies from the father. This means that both parents must be carriers of one or more missing or changed alpha globin genes to have a child who is affected with Hemoglobin H Disease or Alpha-Thalassemia Major.

What do my carrier results mean?

One missing or changed alpha globin gene was identified with your Horizon test. People with one missing or changed alpha globin gene are Alpha-Thalassemia silent carriers. People who are silent carriers for Alpha-Thalassemia usually have no health problems and have normal hemoglobin levels. Thalassemia can occur in people of any ethnicity. It is more common in people with Chinese, Southeast Asian, Indian, Middle Eastern, African, and Mediterranean ancestry.

If your partner is a carrier for Alpha-Thalassemia with two genes missing or changed on the same chromosome (in 'cis'), you would have a 1 in 4, or 25%, chance in each pregnancy of having a child with Hemoglobin H Disease. You are not at risk for having a baby with Alpha-Thalassemia Major. The majority of people of Asian ancestry who have two missing alpha globin genes have them on the same chromosome (in 'cis').

If your partner is a carrier for Alpha-Thalassemia with two genes missing or changed that are located on opposite chromosomes (in "trans), each of your children would have a 50% chance of being carriers of Alpha-Thalassemia (with two genes missing or changed on opposite chromosomes), but you are not at risk to have a child with either Hemoglobin H Disease or Alpha-Thalassemia Major. The majority of people of African-American ancestry who have two missing alpha-globin genes have them on opposite chromosomes.

If your partner is an Alpha-Thalassemia Silent Carrier (with one gene missing or changed), each of your children would have a 25% chance of being carriers of Alpha-Thalassemia (with two genes missing or changed on opposite chromosomes) and a 50% chance of being Alpha-Thalassemia Silent carriers. You would not be at risk to have a child with either Hemoglobin H Disease or Alpha-Thalassemia Major.

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 Alpha-Thalassemia ordered by a health care professional. If your partner is not found to be a carrier for Alpha-Thalassemia, your risk of having a child with Hemoglobin H Disease is greatly reduced. Couples at risk of having a baby with Hemoglobin H Disease 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. If you are not yet pregnant, your partner can have carrier screening for Alpha-Thalassemia ordered by a health care professional. If your partner is found to be a carrier for Alpha-Thalassemia (with two missing or non-working alpha globin genes on the same chromosome, in 'cis') 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 Hemoglobin H Disease
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for Hemoglobin H Disease
- Adoption or use of a sperm or egg donor who is not a carrier for Alpha-Thalassemia

What resources are available?

- March of Dimes: http://www.marchofdimes.org/baby/thalassemia.aspx
- Cooley's Anemia Foundation: www.thalassemia.org
- Prenatal diagnosis done by CVS: http://www.marchofdimes.org/chorionic-villus-sampling.

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CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2

Understanding Your Horizon Carrier Screen Results

What does being a carrier mean?

Your result shows that you are a carrier of congenital dyserythropoietic anemia type 2 (CDA2). 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 CDA2. Further testing can be done to see if your partner or donor is a carrier.

What is congenital dyserythropoietic anemia type 2 (CDA2)?

CDA2 affects the way the body makes red blood cells. People with CDA2 can start to have symptoms any time from before birth to adulthood. Usually, the earlier someone with CDA2 has symptoms, the more severe the condition will be. People with CDA2 have less healthy red blood cells (anemia) than the average person. The anemia can range from mild to severe. People with anemia can be extremely tired, have shortness of breath, headaches, and a rapid or irregular heartbeat. People with CDA2 can also have yellowing of the skin (jaundice), an enlarged liver and spleen, and gallstones. As people with CDA2 age, they can have a buildup of iron in the tissues of the body (iron overload). Iron overload can cause heart disease, diabetes, and liver damage (cirrhosis). People with CDA2 can also have fertility problems and low bone density (thinner or weaker bones). With treatment, people with CDA2 can live into adulthood.[1,2,3,4,5]

Some people with severe symptoms of CDA2 have been treated with stem cell transplantation (SCT) from cord blood or bone marrow.[3,4] Siblings have a higher chance of being a match for SCT than unrelated donors, so cord blood banking could be considered if your children are at risk. More information can be found at: parentsguidecordblood.org/en.

People with CDA2 who do not need SCT have other treatment options. Blood transfusions from donors or medications can be used to remove some of the excess iron in the blood. Babies with CDA2 who have anemia before birth can need blood transfusions during pregnancy. People with CDA2 with severe anemia can also have their spleen removed (splenectomy).[2,3,4] Clinical trials involving potential new treatments for this condition could be available (see clinicaltrials.gov).

What causes congenital dyserythropoietic anemia type 2 (CDA2)?

CDA2 is caused by changes, or variants, in the SEC23B 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 SEC23B gene. Carriers of CDA2 have one working copy and one nonworking copy of the gene. People with CDA2 have no working copies of the gene.

CDA2 is usually passed down, or inherited, from both genetic parents. We inherit one copy of the SEC23B 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 CDA2. Each child also has a 1 in 2 (50%) chance of being a carrier of CDA2 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 congenital dyserythropoietic anemia type 2 (CDA2)?

If your partner or donor also has a nonworking copy of the SEC23B gene, your children could have CDA2. Each child you have together would have a 1 in 4 (25%) chance of having CDA2. 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 SEC23B carrier screening and no variants are found, the chance that your children would have CDA2 is very low. No further testing would usually be needed for you, your partner or donor, or your children related to CDA2.

What can I do next?

If you want to know if your children are at risk for CDA2, your partner or donor would need to have SEC23B 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 CDA2 carrier, your children would be at risk for having CDA2.

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 CDA2. These tests both have a small risk of miscarriage. Babies can also be tested for CDA2 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 CDA2 during pregnancy;
- natural pregnancy and testing the baby after birth for CDA2;
- preimplantation genetic testing (PGT-M) with in vitro fertilization (IVF) to test embryos for CDA2;
- adoption; or
- use of a sperm or egg donor who had no variants found in SEC23B carrier screening.

Where can I find more information?

- Iron Disorders Institute irondisorders.org
- CVS marchofdimes.org/baby/chorionic-villus-sampling
- Amniocentesis marchofdimes.org/pregnancy/amniocentesis

What does this mean for my family?

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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 result so they can decide if they want carrier screening for CDA2.

References

1. MedLinePlus example: MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US).Congenital dyserythropoietic anemia; [updated 2009 Jul 1; cited 2024 Aug 21]; [about 2 p.]. Available from: https://medlineplus.gov/genetics/condition/congenital-dyserythropoietic-anemia/.
2. Musri MM et al. New Cases and Mutations in SEC23B Gene Causing Congenital Dyserythropoietic Anemia Type II. Int J Mol Sci. 2023 Jun 9;24(12):9935. doi: 10.3390/ijms24129935. PMID: 37373084; PMCID: PMC10298408.
3. Iolascon A et al. Congenital dyserythropoietic anemias. Blood. 2020 Sep 10;136(11):1274-1283. doi: 10.1182/blood.2019000948. PMID: 32702750.
4. King R et al. The congenital dyserythropoieitic anemias: genetics and pathophysiology. Curr Opin Hematol. 2022 May 1;29(3):126-136. doi: 10.1097/MOH.0000000000000697. Epub 2021 Dec 24. PMID: 35441598; PMCID: PMC9021540.
5. Hassan MM et al. Congenital Dyserythropoietic Anemia Type II: A Case Report. Cureus. 2022 Aug 12;14(8):e27933. doi: 10.7759/cureus.27933. PMID: 36120266; PMCID: PMC9464459.

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horizon
natural carrier screen

PSEUDOCHOLINESTERASE DEFICIENCY

Understanding Your Horizon Carrier Screen Results

What Is Pseudocholinesterase Deficiency?

Pseudocholinesterase Deficiency (also known as Succinylcholine Sensitivity or Butyrylcholinesterase Deficiency) is an inherited disorder that causes an affected person to become temporarily paralyzed and stop breathing (apnea) when certain medications are used, especially specific muscle relaxants such as succinylcholine (suxamethonium) and mivacurium, which are typically given as part of anesthesia for surgeries. Affected individuals have no symptoms unless they are given these medications. If the medications that cause the paralysis and apnea are avoided, symptoms will not occur. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Pseudocholinesterase Deficiency?

Pseudocholinesterase Deficiency is caused by changes, or mutations, in both copies of the BCHE gene pair. These mutations cause the genes to not work properly or not work at all. When both copies of the BCHE gene are not working correctly and certain anesthesia medications are given, it leads to the symptoms described above. Pseudocholinesterase Deficiency is inherited in an autosomal recessive manner. This means that, in most cases, both parents must be carriers of a mutation in one copy of the BCHE gene to have a child with Pseudocholinesterase Deficiency. People who are carriers for Pseudocholinesterase Deficiency are usually healthy and do not have symptoms, nor do they have Pseudocholinesterase Deficiency 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 Pseudocholinesterase Deficiency, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their BCHE gene mutations to the child, who will then have this condition. Individuals found to carry more than one mutation for Pseudocholinesterase Deficiency should discuss their risk for having an affected child, and any specific risks 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 Pseudocholinesterase Deficiency ordered by a health care professional. If your partner is not found to be a carrier for Pseudocholinesterase Deficiency, your risk of having an affected child is greatly reduced. If your partner is found to be a carrier, you can consider having prenatal diagnostic testing done through chorionic villus sampling (CVS) or amniocentesis during pregnancy to test the fetus for this condition, or can have the baby tested after birth. If you are not yet pregnant, your partner can have carrier screening for Pseudocholinesterase Deficiency ordered by a health care professional. If your partner is found to be a carrier for Pseudocholinesterase Deficiency, you have several reproductive options to consider:

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

What resources are available?

- Genetics Home Reference: https://ghr.nlm.nih.gov/condition/pseudocholinesterase-deficiency
- Prenatal diagnosis by CVS: http://www.marchofdimes.org/chorionic-villus-sampling.aspx
- Prenatal diagnosis by amniocentesis: http://www.marchofdimes.org/amniocentesis.aspx
- Preimplantation genetic diagnosis (PDG) with IVF: http://www.natera.com/spectrum

201 Industrial Road, Suite 410 | San Carlos, CA 94070 | www.natera.com | (855) 288-4872 | Fax: (650) 730-1275

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WILSON DISEASE

Understanding Your Horizon Carrier Screen Results

What Is Wilson Disease?

Wilson Disease is an inherited disorder that causes copper from the diet to build up in certain parts of the body, especially in the liver, eyes, and brain. Signs and symptoms of Wilson Disease usually begin in the teenage years and in rare cases not until adulthood. Symptoms include liver disease, nervous system and psychiatric problems, and specific eye findings called Kayser-Fleischer rings (green/brown colored areas of excess copper on the surface of the eyes that do not interfere with vision). Other symptoms may include problems with coordination, movement, and behavior. Wilson Disease is commonly treated through chelation therapy to remove the excess stored copper from the body. This treatment helps to slow, and in some cases stop, the progression of the disease and improve symptoms. With treatment, people with Wilson Disease can have a normal lifespan. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Wilson Disease?

Wilson Disease is caused by a change, or mutation, in both copies of the ATP7B gene pair. These mutations cause the genes to not work properly or not work at all. Normal function of the ATP7B genes is needed for normal transport of copper within the cells of the body. When both copies of the ATP7B gene do not work correctly, it leads to the symptoms described above. Wilson Disease 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 ATP7B gene to have a child with Wilson Disease. People who are carriers for Wilson Disease are usually healthy and do not have symptoms nor do they have Wilson Disease 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 Wilson Disease there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their ATP7B gene mutations to the child, who will then have the condition. Individuals found to carry more than one mutation for Wilson Disease should discuss their risk for having an affected child, and any potential risks 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 Wilson Disease ordered by a health care professional. If your partner is not found to be a carrier for Wilson Disease, your risk of having a child with this condition is greatly reduced. Couples at risk of having a baby with Wilson Disease 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. If you are not yet pregnant, your partner can have carrier screening for Wilson Disease ordered by a health care professional. If your partner is found to be a carrier for Wilson Disease, you have several reproductive options to consider:

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

What resources are available?

- Genetics Home Reference: http://ghr.nlm.nih.gov/condition/wilson-disease
- 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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VARIANT DETAILS

ATP7B, c.2479C>T (p.R827W), likely pathogenic

- The c.2479C>T (p.R827W) variant in the ATP7B gene has been observed at a frequency of 0.0039% 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 Wilson disease (PMID: 24720933, 33640437, 30232804).
- This variant has been reported in ClinVar [ID: 285881].

BCHE, c.635C>T (p.A212V), likely pathogenic

- The c.635C>T (p.A212V) variant in the BCHE gene has been observed at a frequency of 0.2113% 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 pseudocholinesterase deficiency (PMID: 15731589, 7618741).
- Functional studies demonstrated that this variant causes impaired protein function (PMID: 7618741, 15731589).
- This variant has been reported in ClinVar [ID: 370854].

HBA1/HBA2, alpha 3.7 deletion, pathogenic

- The alpha 3.7 or 4.2 deletion of the HBA1/HBA2 gene is a recombination deletion between the HBA1 and HBA2 gene, resulting in loss of one copy of the HBA1/HBA2 genes.
- Single allele deletion involving one of the four copies of the HBA1/HBA2 genes (alpha 3.7 deletion or alpha 4.2 deletion) has been reported in conjunction with deletions encompassing both HBA1 and HBA2 genes in individuals with HbH disease (PMID: 20301608, 7734346, 27492767, 29032940). Two single allele deletions in trans (alpha 3.7 deletion homozygous, alpha 4.2 deletion in trans, or alpha 3.7 deletion in trans with alpha 4.2 deletion) have been reported in individuals with alpha-thalassemia trait (PMID: 20301608, 29032940).
- This variant has been described in ClinVar [ID: 433555, 648517].

SEC23B, c.40C>T (p.R14W), pathogenic

- The c.40C>T (p.R14W) variant in the SEC23B gene has been observed at a frequency of 0.0237% in the gnomAD v2.1.1 dataset.
- This variant has been reported in a homozygous state or in conjunction with another variant in individual(s) with congenital dyserythropoietic anemia, type II (PMID: 29901818, 29846281, 27471141, 25044164).
- Functional studies demonstrated that this variant causes reduced gene expression (PMID: 19561605).
- This variant has been reported in ClinVar [ID: 1223].

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

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

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

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Directeur scientifique/Chef de laboratoire/Biochimiste Clinique
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Microbiologiste
Dre Véronique Sandekian, PhD, Mcb.A (No licence : 3111)

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D
D-BIFUNCTIONAL PROTEIN DEFICIENCY (*HSD17B4*) negative
DEAFNESS, AUTOSOMAL RECESSIVE 77 (*LOXHD1*) negative
DIHYDROPTERIDINE REDUCTASE (DHPR) DEFICIENCY (*QDPR*) negative
DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY (*DPYD*) 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
FAMILIAL DYSAUTONOMIA (*IKBKAP*) negative
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, PRF1-RELATED (*PRF1*) negative
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STX11-RELATED (*STX11*) negative
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STXBP2-RELATED (*STXBP2*) negative
FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, UNC13D-RELATED (*UNC13D*) negative
FAMILIAL HYPERCHOLESTEROLEMIA, LDLRAP1-RELATED (*LDLRAP1*) negative
FAMILIAL HYPERCHOLESTEROLEMIA, LDLR-RELATED (*LDLR*) negative
FAMILIAL HYPERINSULINISM, ABCC8-RELATED (*ABCC8*) negative
FAMILIAL NEPHROGENIC DIABETES INSIPIDUS, AQP2-RELATED (*AQP2*) negative
FANCONI ANEMIA, GROUP A (*FANCA*) negative
FANCONI ANEMIA, GROUP C (*FANCC*) negative
FANCONI ANEMIA, GROUP D2 (*FANCD2*) negative
FANCONI ANEMIA, GROUP E (*FANCE*) negative
FANCONI ANEMIA, GROUP F (*FANCF*) negative
FANCONI ANEMIA, GROUP G (*FANCG*) negative
FANCONI ANEMIA, GROUP I (*FANCI*) negative
FANCONI ANEMIA, GROUP J (*BRIP1*) negative
FANCONI ANEMIA, GROUP L (*FANCL*) negative
FARBER LIPOGRANULOMATOSIS (*ASAH1*) negative
FOVEAL HYPOPLASIA (*SLC38A8*) negative
FRASER SYNDROME 3, GRIP1-RELATED (*GRIP1*) negative
FRASER SYNDROME, FRAS1-RELATED (*FRAS1*) negative
FRASER SYNDROME, FREM2-RELATED (*FREM2*) negative
FRIEDREICH ATAXIA (*FXN*) negative
FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY (*FBP1*) negative
FUCOSIDOSIS, FUCA1-RELATED (*FUCA1*) negative
FUMARASE DEFICIENCY (*FH*) negative

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

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

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

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

K
KRABBE DISEASE (*GALC*) negative

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Directeur scientifique/Chef de laboratoire/Biochimiste Clinique
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L
LAMELLAR ICHTHYOSIS, TYPE 1 (*TGM1*) negative
LARON SYNDROME (*GHR*) negative
LEBER CONGENITAL AMAUROSIS 2 (*RPE65*) negative
LEBER CONGENITAL AMAUROSIS TYPE AIPL1 (*AIPL1*) negative
LEBER CONGENITAL AMAUROSIS TYPE GUCY2D (*GUCY2D*) negative
LEBER CONGENITAL AMAUROSIS TYPE TULP1 (*TULP1*) negative
LEBER CONGENITAL AMAUROSIS, IQCB1-RELATED/SENIOR-LOKEN SYNDROME 5 (*IQCB1*) negative
LEBER CONGENITAL AMAUROSIS, TYPE CEP290 (*CEP290*) negative
LEBER CONGENITAL AMAUROSIS, TYPE LCA5 (*LCA5*) negative
LEBER CONGENITAL AMAUROSIS, TYPE RDH12 (*RDH12*) negative
LEIGH SYNDROME, FRENCH-CANADIAN TYPE (*LRPPRC*) negative
LETHAL CONGENITAL CONTRACTURE SYNDROME 1 (*GLE1*) negative
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER (*EIF2B5*) negative
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B1-RELATED (*EIF2B1*) negative
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B2-RELATED (*EIF2B2*) negative
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B3-RELATED (*EIF2B3*) negative
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B4-RELATED (*EIF2B4*) negative
LIG4 SYNDROME (*LIG4*) negative
LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 8 (*TRIM32*) negative
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2A (*CAPN3*) negative
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2B (*DYSF*) negative
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2C (*SGCG*) negative
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2D (*SGCA*) negative
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2E (*SGCB*) negative
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2F (*SGCD*) negative
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2I (*FKRP*) negative
LIPOAMIDE DEHYDROGENASE DEFICIENCY (DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY) (*DLD*) negative
LIPOID ADRENAL HYPERPLASIA (*STAR*) negative
LIPOPROTEIN LIPASE DEFICIENCY (*LPL*) negative
LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (*HADHA*) negative
LRAT-RELATED CONDITIONS (*LRAT*) negative
LUNG DISEASE, IMMUNODEFICIENCY, AND CHROMOSOME BREAKAGE SYNDROME (LICS) (*NSMCE3*) negative
LYSINURIC PROTEIN INTOLERANCE (*SLC7A7*) negative

M
MALONYL-COA DECARBOXYLASE DEFICIENCY (*MLYCD*) negative
MAPLE SYRUP URINE DISEASE, TYPE 1A (*BCKDHA*) negative
MAPLE SYRUP URINE DISEASE, TYPE 1B (*BCKDHB*) negative
MAPLE SYRUP URINE DISEASE, TYPE 2 (*DBT*) negative
MCKUSICK-KAUFMAN SYNDROME (*MKKS*) negative
MECKEL SYNDROME 7/NEPHRONOPHTHISIS 3 (*NPHP3*) negative
MECKEL-GRUBER SYNDROME, TYPE 1 (*MKS1*) negative
MECR-RELATED NEUROLOGIC DISORDER (*MECR*) negative
MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (*ACADM*) negative
MEDNIK SYNDROME (*AP1S1*) negative
MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS (*MLC1*) 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 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, MFSDB-RELATED (*MFSD8*) negative
NEURONAL CEROID LIPOFUSCINOSIS, PPT1-RELATED (*PPT1*) negative
NEURONAL CEROID LIPOFUSCINOSIS, TPP1-RELATED (*TPP1*) negative
NGLY1-CONGENITAL DISORDER OF GLYCOSYLATION (*NGLY1*) negative
NIEMANN-PICK DISEASE, TYPE C1 / D (*NPC1*) negative
NIEMANN-PICK DISEASE, TYPE C2 (*NPC2*) negative
NIEMANN-PICK DISEASE, TYPES A / B (*SMPD1*) negative
NIJMEGEN BREAKAGE SYNDROME (*NBN*) negative
NON-SYNDROMIC HEARING LOSS, GJB2-RELATED (*GJB2*) negative
NON-SYNDROMIC HEARING LOSS, MYO15A-RELATED (*MYO15A*) negative
NONSYNDROMIC HEARING LOSS, OTOA-RELATED (*OTOA*) negative
NONSYNDROMIC HEARING LOSS, OTOF-RELATED (*OTOF*) negative
NONSYNDROMIC HEARING LOSS, PJVK-RELATED (*PJVK*) negative
NONSYNDROMIC HEARING LOSS, SYNE4-RELATED (*SYNE4*) negative
NONSYNDROMIC HEARING LOSS, TMC1-RELATED (*TMC1*) negative
NONSYNDROMIC HEARING LOSS, TMPRSS3-RELATED (*TMPRSS3*) negative
NONSYNDROMIC INTELLECTUAL DISABILITY (*CC2D1A*) negative
NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (*SAMD9*) negative

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

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

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

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

S
SALLA DISEASE (*SLC17A5*) **negative**
SANDHOFF DISEASE (*HEXB*) **negative**
SCHIMKE IMMUNOOSSEOUS DYSPLASIA (*SMARCAL1*) **negative**
SCHINDLER DISEASE (*NAGA*) **negative**
SEGAWA SYNDROME, TH-RELATED (*TH*) **negative**
SENIOR-LOKEN SYNDROME 4/NEPHRONOPHTHISIS 4 (*NPHP4*) **negative**
SEPIAPTERIN REDUCTASE DEFICIENCY (*SPR*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3D-RELATED (*CD3D*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3E-RELATED (*CD3E*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), FOXN1-RELATED (*FOXN1*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), IKBKB-RELATED (*IKBKB*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), IL7R-RELATED (*IL7R*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), JAK3-RELATED (*JAK3*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), PTPRC-RELATED (*PTPRC*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), RAG1-RELATED (*RAG1*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY, ADA-Related (*ADA*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY, TYPE ATHABASKAN (*DCLRE1C*) **negative**
SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY (*DYNC2H1*) **negative**
SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED (*SBDS*) **negative**
SIALIDOSIS (*NEU1*) **negative**
SJÖGREN-LARSSON SYNDROME (*ALDH3A2*) **negative**
SMITH-LEMLI-OPITZ SYNDROME (*DHCR7*) **negative**
SPASTIC PARAPLEGIA, TYPE 15 (*ZFYVE26*) **negative**
SPASTIC TETRAPLEGIA, THIN CORPUS CALLOSUM, AND PROGRESSIVE MICROCEPHALY (SPATCCM) (*SLC1A4*) **negative**
SPG11-RELATED CONDITIONS (*SPG11*) **negative**
SPINAL MUSCULAR ATROPHY (*SMN1*) **negative** SMN1: >/= 3 copies; g.27134T>G: absent; the g.27134T>G variant does not modify carrier risk in individuals who carry 3 or more copies of SMN1.
SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS TYPE 1 (*IGHMBP2*) **negative**
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 10 (*ANO10*) **negative**
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12 (*WWOX*) **negative**
SPONDYLOCOSTAL DYSOSTOSIS 1 (*DLL3*) **negative**
SPONDYLOTHORACIC DYSOSTOSIS, MESP2-Related (*MESP2*) **negative**
STEEL SYNDROME (*COL27A1*) **negative**
STEROID-RESISTANT NEPHROTIC SYNDROME (*NPHS2*) **negative**
STUVE-WIEDEMANN SYNDROME (*LIFR*) **negative**
SURF1-RELATED CONDITIONS (*SURF1*) **negative**
SURFACTANT DYSFUNCTION, ABCA3-RELATED (*ABCA3*) **negative**

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

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

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

W
WALKER-WARBURG SYNDROME, CRPPA-RELATED (*CRPPA*) **negative**
WALKER-WARBURG SYNDROME, FKTN-RELATED (*FKTN*) **negative**
WALKER-WARBURG SYNDROME, LARGE1-RELATED (*LARGE1*) **negative**
WALKER-WARBURG SYNDROME, POMT1-RELATED (*POMT1*) **negative**
WALKER-WARBURG SYNDROME, POMT2-RELATED (*POMT2*) **negative**
WARSAW BREAKAGE SYNDROME (*DDX11*) **negative**
WERNER SYNDROME (*WRN*) **negative**
WILSON DISEASE (*ATP7B*) **see first page**
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**

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horizon
natera carrier screen

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

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

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

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

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

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

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

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

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

Friedreich Ataxia (FXN)

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

Friedreich Ataxia Repeat Categories

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

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natera carrier screen

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/genes/option/ft.pdf 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.

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Légende: **AN** = anormal **AB** = anormal bas **AH** = anormal haut **C** = critique **CB** = critique bas **CH** = critique haut **X** = absurde **XB** = absurde bas **XH** = absurde haut

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