Patient Information		Test Information	
Patient Name:		Ordering Physician:	Coralie Beauchamps
		Clinic Information:	Clinique Ovo
Date Of Birth:			
Gender:			
Ethnicity:		Phone:	
		Report Date:	07/26/2025
Patient ID:	N/A	Sample Collected:	07/11/2025
Medical Record #:	N/A	Sample Received:	07/14/2025
Collection Kit:		Sample Type:	Blood
Accession ID:			
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:

CARRIER for Shwachman-Diamond Syndrome, SBDS-Related

Positive for the pathogenic variant c.258+2T>C in the SBDS gene. If this individual's partner is a carrier for SHWACHMAN-DIAMOND SYNDROME, SBDS-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 Spinal Muscular Atrophy

Positive for one copy of the SMN1 gene. Negative for the g.27134T>G variant; this finding does not change or modify this individual's carrier status. If this individual's partner is a carrier for Spinal Muscular Atrophy, their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

Negative for 549 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-policy/h-all/. Please see the following pages of this report for a comprehensive list of all conditions included on this individual's screen.

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

RECOMMENDATIONS

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

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

Linyan Meng, Ph.D
Laboratory Director, Baylor Genetics

J. Dunn Hamilton, Ph.D, FACMGG
Senior Laboratory Director, Natera

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

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Dr Artak Tadevosyan, PhD, DEPD, CSPQ, FCACB (No licence : 2014-199)

Microbiologiste
Dre Véronique Sandekian, PhD, Mcb.A (No licence : 3111)

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horizon
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SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED

Understanding Your Horizon Carrier Screen Results

What Is Shwachman-Diamond Syndrome, SBDS-Related?

Shwachman-Diamond Syndrome, SBDS-Related is an inherited disorder that affects the bones, the bone marrow, and the pancreas. Bone abnormalities are commonly seen in the hips and knees and sometimes include short ribs and narrow rib cage, which can lead to serious breathing problems. Affected individuals have slow bone growth and short stature. The bone marrow problems in Shwachman-Diamond Syndrome, SBDS-Related include lower production of white blood cells that fight infections (called neutropenia), causing an increased numbers of infections. Some affected individuals also have a reduced number of red blood cells, which leads to anemia and less oxygen getting to the cells of the body; and some have a reduced number of platelets, which leads to easy bruising and prolonged bleeding. The bone marrow problems also increase the risk for a type of cancer called Acute Myeloid Leukemia (AML). Individuals with Shwachman-Diamond Syndrome, SBDS-Related do not make enough digestive enzymes in their pancreas (called pancreatic insufficiency) and have trouble digesting their food. If not treated, this causes slow growth, lack of weight gain, and malnutrition. Symptoms sometimes also include problems with the heart, eyes, teeth, skin, and endocrine system. Some affected children have delayed development and speech. Currently there is no cure for this disorder and treatment includes daily supplementation with pancreatic enzymes and doctor-prescribed vitamins along with other medical management based on symptoms. 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 Shwachman-Diamond Syndrome, SBDS-Related?

Shwachman-Diamond Syndrome, SBDS-Related is caused by changes, or mutations, in both copies of the SBDS gene pair. These mutations cause the genes to not work properly or not work at all. When both copies of the SBDS gene do not work correctly it leads to the symptoms described above. Shwachman-Diamond Syndrome, SBDS-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 SBDS gene to have a child with Shwachman-Diamond Syndrome, SBDS-Related. People who are carriers for Shwachman-Diamond Syndrome, SBDS-Related are usually healthy and do not have symptoms, nor do they have Shwachman-Diamond Syndrome, SBDS-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 Shwachman-Diamond Syndrome, SBDS-Related, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their SBDS gene mutations to the child, who will then have this disorder. Individuals found to carry more than one mutation for Shwachman-Diamond Syndrome, SBDS-Related should discuss their risk for having an affected child, and any specific risks to their own health, with their health care provider. There are a number of other forms of Shwachman-Diamond Syndrome, SBDS-Related, each caused by mutations in different genes. A person who is a carrier of a mutation in the SBDS gene 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 Shwachman-Diamond Syndrome, SBDS-Related ordered by a health care professional. If your partner is not found to be a carrier for Shwachman-Diamond Syndrome, SBDS-Related, 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, or you can have the baby tested after birth for this disorder. If you are not yet pregnant, your partner can have carrier screening for Shwachman-Diamond Syndrome, SBDS-Related ordered by a health care professional. If your partner is found to be a carrier for Shwachman-Diamond Syndrome, SBDS-Related, 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 Shwachman-Diamond Syndrome, SBDS-Related
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for Shwachman-Diamond Syndrome, SBDS-Related
- Adoption or use of a sperm or egg donor who is not a carrier for Shwachman-Diamond Syndrome, SBDS-Related

What resources are available?

- Genetics Home Reference: https://ghr.nlm.nih.gov/condition/shwachman-diamond-syndrome
- GeneReviews: http://www.ncbi.nlm.nih.gov/books/NBK1256/
- 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

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SPINAL MUSCULAR ATROPHY

Understanding Your Horizon Carrier Screen Results

What Is Spinal Muscular Atrophy?

Spinal Muscular Atrophy (SMA) is a serious inherited disorder that typically begins in infancy or childhood and causes worsening muscle weakness, decreased ability to breathe, and loss of motor skills. Most children with SMA have one of the early-onset forms with symptoms that begin in infancy. Without treatment, death often occurs before the age of two. Some children have juvenile-onset SMA and develop muscle weakness and other symptoms later in childhood and typically have a normal lifespan. In rare cases symptoms do not begin until early adulthood, are less severe, and do not affect lifespan. Currently there is no cure for SMA, although some affected individuals may benefit from new medications that can lessen or stop the progression of symptoms, especially when treatment is started early. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Spinal Muscular Atrophy?

SMA is caused by a change, or mutation, in both copies the SMN1 gene pair. These mutations, which often delete part or all of the gene, cause the genes to work improperly or not work at all. When both copies of the SMN1 gene are missing or do not work correctly, it leads to the symptoms described above. SMA 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 SMN1 gene to have a child with SMA. People who are carriers are usually healthy and do not have symptoms nor do they have SMA themselves. Usually a child inherits two copies of each gene, one from their mother and one from their father. If the mother and father are found to be SMA carriers, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their SMN1 gene mutations to the child, who would then have SMA. With further testing (not offered through Natera), it is sometimes, but not always, possible to determine whether a given carrier couple is at risk to have a child with a severe, early-onset form of SMA, the juvenile form, or the later-onset form. Individuals found to carry more than one mutation for SMA 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 positive SMA results. A genetic counselor in your region 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 genetic marker. You are encouraged to inform your family members of your test results as they may wish to consider being tested for SMA carrier status themselves. If you are pregnant, your partner can have carrier screening for SMA ordered by a health care professional. Partner screening may include SMN1 testing and possibly Enhanced SMA testing. Enhanced SMA testing can provide information on the chance to still be a carrier even after a normal (negative) SMA carrier screen. Your doctor or a local genetic counselor can help decide which carrier test is best for your partner. If your partner is not found to be a carrier of SMA, your risk of having a child with SMA is greatly reduced. Couples at risk of having a baby with SMA can opt to have prenatal diagnosis done through chorionic villus sampling or amniocentesis during pregnancy or can choose to have the baby tested after birth for SMA. If you are not yet pregnant, your partner can have carrier testing for SMA ordered by a health care professional. Partner testing may include SMN1 testing and possibly Enhanced SMA testing. Enhanced SMA testing can provide information on the chance to still be a carrier even after a normal (negative) SMA carrier screen. Your doctor or a genetic counselor can help decide which carrier test is best for your partner. If your partner is found to be a carrier for SMA, 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 SMA
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos for SMA
- Adoption or use of a sperm or egg donor who is not a carrier for SMA

What resources are available?

- Families of SMA: www.curesma.org
- GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1352/
- Prenatal diagnosis done by CVS: http://www.marchofdimes.org/chorionic-villus-sampling.aspx
- Prenatal diagnosis done by amniocentesis: http://www.marchofdimes.org/amniocentesis.aspx
- PGD with IVF: http://natera.com/spectrum

201 Industrial Road, Suite 410 | San Carlos, CA 94070 | www.natera.com | 650-249-9090 | Fax: 1 650-730-3172

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VARIANT DETAILS

SBDS, c.258+2T>C, pathogenic

- The c.258+2T>C variant in the SBDS gene has been observed at a frequency of 0.3879% in the gnomAD v2.1.1 dataset.
- This variant has been previously reported in a homozygous state or in conjunction with another variant in individuals with Shwachman-Diamond syndrome (PMID: 12496757, 14749921, 15860664, 22935661).
- 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 been reported in ClinVar [ID: 3196].

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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*) **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 (*CTSD*) **negative**
COHEN SYNDROME (*VPS13B*) **negative**
COL11A2-RELATED CONDITIONS (*COL11A2*) **negative**
COMBINED MALONIC AND METHYLMALONIC ACIDURIA (*ACSF3*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (*GFM1*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (*TSFM*) **negative**
COMBINED PITUITARY HORMONE DEFICIENCY 1 (*POU1F1*) **negative**
COMBINED PITUITARY HORMONE DEFICIENCY-2 (*PROP1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (*CYP11B1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (*CYP17A1*) **negative**
CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (*CYP21A2*) **negative**
CONGENITAL ADRENAL INSUFFICIENCY, CYP11A1-RELATED (*CYP11A1*) **negative**
CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (*MPL*) **negative**
CONGENITAL CHRONIC DIARRHEA (*DGAT1*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, ALG1-RELATED (*ALG1*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-Related (*PMM2*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (*MPI*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (*ALG6*) **negative**
CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (*SEC23B*) **negative**
CONGENITAL FINNISH NEPHROSIS (*NPHS1*) **negative**
CONGENITAL HYDROCEPHALUS 1 (*CCDC88C*) **negative**
CONGENITAL HYPERINSULINISM, KCNJ11-Related (*KCNJ11*) **negative**
CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (CIPA) (*NTRK1*) **negative**
CONGENITAL MYASTHENIC SYNDROME, CHAT-RELATED (*CHAT*) **negative**
CONGENITAL MYASTHENIC SYNDROME, CHRNE-RELATED (*CHRNE*) **negative**
CONGENITAL MYASTHENIC SYNDROME, 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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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

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

Directeur scientifique/Chef de laboratoire/Biochimiste Clinique
Dr Artak Tadevosyan, PhD, DEPD, CSPQ, FCACB (No licence : 2014-199)

Microbiologiste
Dre Véronique Sandekian, PhD, Mcb.A (No licence : 3111)

Patient Information
Patient Name:

Test Information
Ordering Physician:

Clinic Information:

Date Of Birth:
Case File ID:

Report Date:

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, 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
NUMEGEN 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

201 Industrial Road Suite 410 | San Carlos CA 94070 | www.natera.com | 650-249 9090 | Fax 1 650-730 1273

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Directeur scientifique/Chef de laboratoire/Biochimiste Clinique
Dr Artak Tadevosyan, PhD, DEPD, CSPQ, FCACB (No licence : 2014-199)

Microbiologiste
Dre Véronique Sandekian, PhD, Mcb.A (No licence : 3111)

Patient Information
Patient Name:

Test Information
Ordering Physician:

Clinic Information:

Date Of Birth:
Case File ID:

Report Date:

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*) **negative**
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*) **see first page**
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*) **see first page**
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*) **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**

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Directeur scientifique/Chef de laboratoire/Biochimiste Clinique
Dr Artak Tadevosyan, PhD, DEPD, CSPQ, FCACB (No licence : 2014-199)

Microbiologiste
Dre Véronique Sandekian, PhD, Mcb.A (No licence : 3111)

Patient Information
Patient Name:

Date Of Birth:
Case File ID:

Test Information
Ordering Physician:

Clinic Information:

Report Date:

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Z
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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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/panels-optical/b.zip/ 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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