Patient Information		Test Information	
Patient Name:		Ordering Physician:	Coralie Beauchamps
		Clinic Information:	Clinique Ovo
Date Of Birth:			
Gender:	Male		
Patient ID:	N/A	Phone:	
Medical Record #:	N/A	Report Date:	07/22/2026
Collection Kit:		Sample Collected:	
Accession ID:		Sample Received:	
Case File ID:		Sample Type:	
Ethnicity:			

natera | Horizon™

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 GLYCOGEN STORAGE DISEASE TYPE 5 (McArdle Disease)

Positive for the pathogenic variant c.148C>T (p.R50*) in the PYGM gene. If this individual's partner is a carrier for GLYCOGEN STORAGE DISEASE TYPE 5 (McArdle Disease), 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 550 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/carrier-screening/horizon/. 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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Laboratory Director, Baylor Genetics

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Senior Laboratory Director, Natera

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Laboratory Director, Natera

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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	Test Information	
Patient Name:	Ordering Physician:	Coralie Beauchamps
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GLYCOGEN STORAGE DISEASE TYPE 5 (McArdle Disease)

Understanding Your Horizon Carrier Screen Results

What is Glycogen Storage Disease, Type 5 (McArdle Disease)?

Glycogen Storage Disease, Type 5 (GSD5), also called McArdle disease, is an inherited disorder in which the body cannot change glycogen to glucose, the sugar used by the body for energy. This leads lack of enough energy for muscle cells to work properly. The symptoms of GSD5 can vary from mild to severe and commonly occur in young adults between the ages of 20 and 30. Symptoms include muscle cramping, pain, weakness, soreness, and fatigue when exercising. Individuals with this disorder may also experience blood in the urine and temporary kidney failure. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Glycogen Storage Disease Type 5 (McArdle Disease)?

Glycogen Storage Disease, Type 5 (GSD5) is caused by a change, or mutation, in both copies of the PYGM gene pair. These mutations cause the genes to not work properly or not work at all. When both copies of this gene do not work correctly, it leads to the symptoms described above. GSD5 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 PYGM gene to have a child with GSD5. People who are carriers for GSD5 are usually healthy and do not have symptoms nor do they have GSD5 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 GSD5 there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their PYGM gene mutations to the child, who will then have GSD5. Individuals found to carry more than one mutation for GSD5 should discuss their risk for having an affected child and any potential effects to their own health with their health care provider. There are a number of other subtypes of Glycogen Storage Disease, each caused by mutations in different genes. A person who is a carrier for GSD5 is not likely to be at significantly increased risk for having a child with other subtypes of this disorder.

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 GSD5 ordered by a health care professional. If your partner is not found to be a carrier for GSD5, your risk of having a child with this condition is greatly reduced. Couples at risk of having a baby with GSD5 can opt to have prenatal diagnosis done through chorionic villus sampling (CVS) or amniocentesis during pregnancy or can choose to have the baby tested after birth for GSD5. If you are not yet pregnant, your partner can have carrier screening for GSD5 ordered by a health care professional. If your partner is found to be a carrier for GSD5 you have several reproductive options to consider:

- Natural pregnancy with or without prenatal diagnosis of the fetus or testing the baby after birth for Glycogen Storage Disease, Type 5
- Preimplantation genetic diagnosis (PGD) with in vitro fertilization (IVF) to test embryos Glycogen Storage Disease, Type 5
- Adoption or use of a sperm or egg donor who is not a carrier for Glycogen Storage Disease, Type 5

What resources are available?

- Genetics Home Reference: http://ghr.nlm.nih.gov/condition/glycogen-storage-disease-type-v
- 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

201 Industrial Road, Suite 410 | San Carlos, CA 94070 | www.natera.com | 650-249-9090 | Fax 1-650-730-2272

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VARIANT DETAILS

PYGM, c.148C>T (p.R50*), pathogenic

- The c.148C>T (p.R50*) variant in the PYGM gene has been observed at a frequency of 0.1499% in the gnomAD v2.1.1 dataset.
- This variant has been reported in a homozygous state or with another variant in individual(s) with glycogen storage disease, type V (PMID: 8316268, 16786513, 21802952, 22250184, 23653251).
- Functional studies demonstrated that this variant causes impaired protein function (PMID: 22730558).
- This premature termination variant is predicted to cause nonsense-mediated decay (NMD) in a gene where loss-of-function is a known mechanism of disease.
- This variant has been reported in ClinVar [ID: 2298].

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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-GOUTIERES 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-GOUTIERES 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**

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C
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

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*) see first page
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

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H
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-GRASBECK 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

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

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M
METHYLMALONIC ACIDURIA, MMAA-RELATED (*MMAA*) **negative**
METHYLMALONIC ACIDURIA, MMAB-RELATED (*MMAB*) **negative**
METHYLMALONIC ACIDURIA, TYPE MUT(0) (*MUT*) **negative**
MEVALONIC KINASE DEFICIENCY (*MVK*) **negative**
MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II (*PCNT*) **negative**
MICROPHTHALMIA / ANOPHTHALMIA, VSX2-RELATED (*VSX2*) **negative**
MITOCHONDRIAL COMPLEX 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**
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 LEFEVRE SYNDROME (*CTSC*) **negative**
PARKINSON DISEASE 15 (*FBXO7*) **negative**
PENDRED SYNDROME (*SLC26A4*) **negative**
PERLMAN SYNDROME (*DIS3L2*) **negative**
PGM3-CONGENITAL DISORDER OF GLYCOSYLATION (*PGM3*) **negative**
PHENYLKETONURIA (*PAH*) **negative**
PIGN-CONGENITAL DISORDER OF GLYCOSYLATION (*PIGN*) **negative**
PITUITARY HORMONE DEFICIENCY, COMBINED 3 (*LHX3*) **negative**
POLG-RELATED DISORDERS (*POLG*) **negative**
POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE (*PKHD1*) **negative**
PONTOCEREBELLAR HYPOPLASIA, EXOSC3-RELATED (*EXOSC3*) **negative**
PONTOCEREBELLAR HYPOPLASIA, RARS2-RELATED (*RARS2*) **negative**
PONTOCEREBELLAR HYPOPLASIA, TSEN2-RELATED (*TSEN2*) **negative**
PONTOCEREBELLAR HYPOPLASIA, TSEN54-RELATED (*TSEN54*) **negative**
PONTOCEREBELLAR HYPOPLASIA, TYPE 1A (*VRK1*) **negative**
PONTOCEREBELLAR HYPOPLASIA, TYPE 2D (*SEPSECS*) **negative**
PONTOCEREBELLAR HYPOPLASIA, VPS53-RELATED (*VPS53*) **negative**
PRIMARY CILIARY DYSKINESIA, CCDC103-RELATED (*CCDC103*) **negative**
PRIMARY CILIARY DYSKINESIA, CCDC39-RELATED (*CCDC39*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAH11-RELATED (*DNAH11*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAH5-RELATED (*DNAH5*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAI1-RELATED (*DNAI1*) **negative**
PRIMARY CILIARY DYSKINESIA, DNAI2-RELATED (*DNAI2*) **negative**
PRIMARY CONGENITAL GLAUCOMA/PETERS ANOMALY (*CYP1B1*) **negative**
PRIMARY HYPEROXALURIA, TYPE 1 (*AGXT*) **negative**
PRIMARY HYPEROXALURIA, TYPE 2 (*GRHPR*) **negative**
PRIMARY HYPEROXALURIA, TYPE 3 (*HOGA1*) **negative**
PRIMARY MICROCEPHALY 1, AUTOSOMAL RECESSIVE (*MCPH1*) **negative**
PROGRESSIVE EARLY-ONSET ENCEPHALOPATHY WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM (*TBCD*) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, ABCB4-RELATED (*ABCB4*) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 1 (PFIC1) (*ATP8B1*) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 2 (*ABCB11*) **negative**
PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 4 (PFIC4) (*TJP2*) **negative**
PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA (*CCN6*) **negative**
PROLIDASE DEFICIENCY (*PEPD*) **negative**
PROPIONIC ACIDEMIA, PCCA-RELATED (*PCCA*) **negative**
PROPIONIC ACIDEMIA, PCCB-RELATED (*PCCB*) **negative**
PSEUDOCHOLINESTERASE DEFICIENCY (*BCHE*) **negative**

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P
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
SJOGREN-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: present; 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*) 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

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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)

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Test Information
Ordering Physician:

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Z
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-CRYL1 critical region is included.

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

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

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

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

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Friedreich Ataxia (FXN)

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

Friedreich Ataxia Repeat Categories

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

Spinal Muscular Atrophy (SMN1)

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

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

Variant Classification

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

Negative Results

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

Additional Comments

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

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