



Donor 7274

Genetic Testing Summary

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

Last Updated: 08/22/25

Donor Reported Ancestry: German, Swedish, Norwegian

Jewish Ancestry: No

| Genetic Test* | Result | Comments/ Donor's Residual Risk** |
|---|---|---|
| Chromosome analysis (karyotype) | Normal male karyotype | No evidence of clinically significant chromosome abnormalities |
| Hemoglobin evaluation | Normal hemoglobin fractionation and MCV/MCH results | Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies |
| Expanded Genetic Disease Carrier Screening Panel attached- 549 diseases by gene sequencing. | Negative for genes sequenced. | |
| Special Testing | | |
| Gene(s): PIEZO1 | Negative through gene sequencing with deletion and duplication analysis | |

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

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

Patient Information

Patient Name: Donor 7274
Date Of Birth: [REDACTED]
Gender: Male
Ethnicity: Northern European
Caucasian
Patient ID: N/A
Medical Record #: 7274-[REDACTED]
Collection Kit: [REDACTED]
Accession ID: N/A
Case File ID: [REDACTED]

Test Information

Ordering Physician: [REDACTED]
Clinic Information: Fairfax Cryobank
Phone: [REDACTED]
Report Date: 05/19/2024
Sample Collected: 05/07/2024
Sample Received: 05/08/2024
Sample Type: Blood

CARRIER SCREENING REPORT

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

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

FINAL RESULTS SUMMARY:**Negative for 549 out of 549 diseases**


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


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


RECOMMENDATIONS

Individuals who would like to review their Horizon report with a Natera Laboratory Genetic Counselor may schedule a telephone genetic information session by calling 650-249-9090 or visiting naterasession.com. Clinicians with questions may contact Natera at 650-249-9090 or email support@natera.com.


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Patient Information

Patient Name: Donor 7274

Test Information

Ordering Physician: [REDACTED]



Clinic Information: Fairfax Cryobank

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: 05/19/2024

DISEASES SCREENED

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

Autosomal Recessive

- 1**
17-BETA HYDROXYSTEROID DEHYDROGENASE 3 DEFICIENCY (*HSD17B3*) **negative**
- 3**
3-BETA-HYDROXYSTEROID DEHYDROGENASE TYPE II DEFICIENCY (*HSD3B2*) **negative**
3-HYDROXY-3-METHYLGLUTARYL-COENZYME A LYASE DEFICIENCY (*HMGCL*) **negative**
3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (*HADH*) **negative**
3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY (*MCCC2*) **negative**
3-PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY (*PHGDH*) **negative**
- 5**
5-ALPHA-REDUCTASE DEFICIENCY (*SRD5A2*) **negative**
- 6**
6-PYRUVYL-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-GOUTIÈRES SYNDROME, RNASEH2A-RELATED (*RNASEH2A*) **negative**
AICARDI-GOUTIÈRES SYNDROME, RNASEH2B-RELATED (*RNASEH2B*) **negative**
AICARDI-GOUTIÈRES SYNDROME, RNASEH2C-RELATED (*RNASEH2C*) **negative**
AICARDI-GOUTIÈRES SYNDROME, TREX1-RELATED (*TREX1*) **negative**
ALPHA-MANNOSIDOSIS (*MAN2B1*) **negative**
ALPHA-THALASSEMIA (*HBA1/HBA2*) **negative**
ALPORT SYNDROME, COL4A3-RELATED (*COL4A3*) **negative**
ALPORT SYNDROME, COL4A4-RELATED (*COL4A4*) **negative**
ALSTROM SYNDROME (*ALMS1*) **negative**
AMISH INFANTILE EPILEPSY SYNDROME (*ST3GAL5*) **negative**
ANDERMANN SYNDROME (*SLC12A6*) **negative**
ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (*GATM*) **negative**
ARGININEMIA (*ARG1*) **negative**
ARGININOSUCCINATE LYASE DEFICIENCY (*ASL*) **negative**
AROMATASE DEFICIENCY (*CYP19A1*) **negative**
ASPARAGINE SYNTHETASE DEFICIENCY (*ASNS*) **negative**
ASPARTYLGLYCOSAMINURIA (*AGA*) **negative**
ATAXIA WITH VITAMIN E DEFICIENCY (*TTPA*) **negative**
ATAXIA-TELANGIECTASIA (*ATM*) **negative**
ATAXIA-TELANGIECTASIA-LIKE DISORDER 1 (*MRE11*) **negative**
ATRANSFERRINEMIA (*TF*) **negative**
AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (*SLC35A3*) **negative**
AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (*AIRE*) **negative**
AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (*ARCI*), SLC27A4-RELATED (*SLC27A4*) **negative**
AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (*SACS*) **negative**
- B**
BARDET-BIEDL SYNDROME, ARL6-RELATED (*ARL6*) **negative**
BARDET-BIEDL SYNDROME, BBS10-RELATED (*BBS10*) **negative**
BARDET-BIEDL SYNDROME, BBS12-RELATED (*BBS12*) **negative**
BARDET-BIEDL SYNDROME, BBS1-RELATED (*BBS1*) **negative**
BARDET-BIEDL SYNDROME, BBS2-RELATED (*BBS2*) **negative**
BARDET-BIEDL SYNDROME, BBS4-RELATED (*BBS4*) **negative**
BARDET-BIEDL SYNDROME, BBS5-RELATED (*BBS5*) **negative**
BARDET-BIEDL SYNDROME, BBS7-RELATED (*BBS7*) **negative**
BARDET-BIEDL SYNDROME, BBS9-RELATED (*BBS9*) **negative**
BARDET-BIEDL SYNDROME, TTC8-RELATED (*TTC8*) **negative**
BARE LYMPHOCYTE SYNDROME, CIITA-RELATED (*CIITA*) **negative**
BARTTER SYNDROME, BSND-RELATED (*BSND*) **negative**
BARTTER SYNDROME, KCNJ1-RELATED (*KCNJ1*) **negative**
BARTTER SYNDROME, SLC12A1-RELATED (*SLC12A1*) **negative**
BATTEN DISEASE, CLN3-RELATED (*CLN3*) **negative**
BETA-HEMOGLOBINOPATHIES (*HBB*) **negative**
BETA-KETOTHIOLASE DEFICIENCY (*ACAT1*) **negative**
BETA-MANNOSIDOSIS (*MANBA*) **negative**
BETA-UREIDOPROPIONASE DEFICIENCY (*UPB1*) **negative**
BILATERAL FRONTOPARIETAL POLYMICROGYRIA (*GPR56*) **negative**
- BIOTINIDASE DEFICIENCY (*BTD*) **negative**
BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBGD) (*SLC19A3*) **negative**
BLOOM SYNDROME (*BLM*) **negative**
BRITTLE CORNEA SYNDROME 1 (*ZNF469*) **negative**
BRITTLE CORNEA SYNDROME 2 (*PRDM5*) **negative**
- C**
CANAVAN DISEASE (*ASPA*) **negative**
CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (*CPS1*) **negative**
CARNITINE DEFICIENCY (*SLC22A5*) **negative**
CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY (*CPT1A*) **negative**
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (*CPT2*) **negative**
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (*SLC25A20*) **negative**
CARPENTER SYNDROME (*RAB23*) **negative**
CARTILAGE-HAIR HYPOPLASIA (*RMRP*) **negative**
CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (*CASQ2*) **negative**
CD59-MEDIATED HEMOLYTIC ANEMIA (*CD59*) **negative**
CEP152-RELATED MICROCEPHALY (*CEP152*) **negative**
CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA (CEDNIK) SYNDROME (*SNAP29*) **negative**
CEREBROTENDINOUS XANTHOMATOSIS (*CYP27A1*) **negative**
CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE C (*PLEKHG5*) **negative**
CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (*NDRG1*) **negative**
CHEDIAK-HIGASHI SYNDROME (*LYST*) **negative**
CHOREOACANTHOCYTOSIS (*VPS13A*) **negative**
CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (*CYBA*) **negative**
CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (*NCF2*) **negative**
CILIOPATHIES, RPGRIP1L-RELATED (*RPGRIP1L*) **negative**
CITRIN DEFICIENCY (*SLC25A13*) **negative**
CITRULLINEMIA, TYPE 1 (*ASS1*) **negative**
CLN10 DISEASE (*CTSD*) **negative**
COHEN SYNDROME (*VPS13B*) **negative**
COL11A2-RELATED CONDITIONS (*COL11A2*) **negative**
COMBINED MALONIC AND METHYLMALONIC ACIDURIA (*ACSF3*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (*GF1M*) **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, RAPSIN-RELATED (*RAPSIN*) **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-METHYLGUTACONIC ACIDURIA, TYPE 3) (*OPA3*) **negative**
CRB1-RELATED RETINAL DYSTROPHIES (*CRB1*) **negative**
CYSTIC FIBROSIS (*CFTR*) **negative**
CYSTINOSIS (*CTNS*) **negative**
CYTOCHROME C OXIDASE DEFICIENCY, PET100-RELATED (*PET100*) **negative**
CYTOCHROME P450 OXIOREDUCTASE DEFICIENCY (*POR*) **negative**
- D**
D-BIFUNCTIONAL PROTEIN DEFICIENCY (*HSD17B4*) **negative**

Patient Information

Patient Name:

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D

DEAFNESS, AUTOSOMAL RECESSIVE 77 (*LOXHD1*) **negative**
 DIHYDROPTERIDINE REDUCTASE (DHPR) DEFICIENCY (*QDPR*) **negative**
 DONNAI-BARROW SYNDROME (*LRP2*) **negative**
 DUBIN-JOHNSON SYNDROME (*ABCC2*) **negative**
 DYSKERATOSIS CONGENITA SPECTRUM DISORDERS (*TERT*) **negative**
 DYSKERATOSIS CONGENITA, RTTEL1-RELATED (*RTTEL1*) **negative**
 DYSTROPHIC EPIDERMOLYSIS BULLOSA, COL7A1-Related (*COL7A1*) **negative**

E

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

F

FACTOR XI DEFICIENCY (*F11*) **negative**
 FAMILIAL DYSAUTONOMIA (*IKBKAP*) **negative**
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, PRF1-RELATED (*PRF1*) **negative**
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STX11-RELATED (*STX11*) **negative**
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STXBP2-RELATED (*STXBP2*) **negative**
 FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, UNC13D-RELATED (*UNC13D*) **negative**
 FAMILIAL HYPERCHOLESTEROLEMIA, LDLRAP1-RELATED (*LDLRAP1*) **negative**
 FAMILIAL HYPERCHOLESTEROLEMIA, LDLR-RELATED (*LDLR*) **negative**
 FAMILIAL HYPERINSULINISM, ABCC8-RELATED (*ABCC8*) **negative**
 FAMILIAL NEPHROGENIC DIABETES INSIPIDUS, AQP2-RELATED (*AQP2*) **negative**
 FANCONI ANEMIA, GROUP A (*FANCA*) **negative**
 FANCONI ANEMIA, GROUP C (*FANCC*) **negative**
 FANCONI ANEMIA, GROUP D2 (*FANCD2*) **negative**
 FANCONI ANEMIA, GROUP E (*FANCE*) **negative**
 FANCONI ANEMIA, GROUP F (*FANCF*) **negative**
 FANCONI ANEMIA, GROUP G (*FANCG*) **negative**
 FANCONI ANEMIA, GROUP I (*FANCI*) **negative**
 FANCONI ANEMIA, GROUP J (*BRIP1*) **negative**
 FANCONI ANEMIA, GROUP L (*FANCL*) **negative**
 FARBER LIPOGRANULOMATOSIS (*ASAH1*) **negative**
 FOVEAL HYPOPLASIA (*SLC38A8*) **negative**
 FRASER SYNDROME 3, GRIP1-RELATED (*GRIP1*) **negative**
 FRASER SYNDROME, FRAS1-RELATED (*FRAS1*) **negative**
 FRASER SYNDROME, FREM2-RELATED (*FREM2*) **negative**
 FRIEDREICH ATAXIA (*FXN*) **negative**
 FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY (*FBP1*) **negative**
 FUCOSIDOSIS, FUCA1-RELATED (*FUCA1*) **negative**
 FUMARASE DEFICIENCY (*FH*) **negative**

G

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

GRACILE SYNDROME (*BCS1L*) **negative**GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY (*GAMT*) **negative****H**

HARLEQUIN ICHTHYOSIS (*ABCA12*) **negative**
 HEME OXYGENASE 1 DEFICIENCY (*HMOX1*) **negative**
 HEMOCHROMATOSIS TYPE 2A (*HFE2*) **negative**
 HEMOCHROMATOSIS, TYPE 3, TFR2-Related (*TFR2*) **negative**
 HEPATOCEREBRAL MITOCHONDRIAL DNA DEPLETION SYNDROME, MPV17-RELATED (*MPV17*) **negative**
 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 (*HLC5*) **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 (*JNV5*) **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, AH11-RELATED (*AH11*) **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/LARYNGOOXYCHOCUTANEOUS SYNDROME, LAMA3-RELATED (*LAMA3*) **negative**

KKRABBE DISEASE (*GALC*) **negative****L**LAMELLAR ICHTHYOSIS, TYPE 1 (*TGM1*) **negative**

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L
 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) (*DLI*) **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 OSTEOPLASTIC PRIMORDIAL DWARFISM TYPE II (*PCNT*) **negative**
 MICROPTHALMIA / ANOPHTHALMIA, VSX2-RELATED (*VSX2*) **negative**
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, ACAD9-RELATED (*ACAD9*) **negative**
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUF56-RELATED (*NDUF56*) **negative**
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUF54-RELATED (*NDUF54*) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 10 (*NDUF52*) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 17 (*NDUF56*) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 19 (*FOXRED1*) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 3 (*NDUF57*) **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 (*MOC52*) **negative**
 MOLYBDENUM COFACTOR DEFICIENCY, TYPE A (*MOC51*) **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) (*AGLU*) **negative**
 MUCOPOLYSACCHARIDOSIS, TYPE III C (SANFILIPPO C) (*HGSNAT*) **negative**
 MUCOPOLYSACCHARIDOSIS, TYPE III D (SANFILIPPO D) (*GNS*) **negative**
 MUCOPOLYSACCHARIDOSIS, TYPE VI (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-ACETYLGUTAMATE 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, PJK-RELATED (*PJK*) **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 IV (*SLC45A2*) **negative**
 OCULOCUTANEOUS ALBINISM TYPE, III (*TYRP1*) **negative**
 OCULOCUTANEOUS ALBINISM, OCA2-RELATED (*OCA2*) **negative**
 OCULOCUTANEOUS ALBINISM, TYPES 1A AND 1B (*TYR*) **negative**
 ODONTO-ONYCHO-DERMAL DYSPLASIA / SCHOPF-SCHULZ-PASSARGE SYNDROME (*WNT10A*) **negative**
 OMENN SYNDROME, RAG2-RELATED (*RAG2*) **negative**
 ORNITHINE AMINOTRANSFERASE DEFICIENCY (*OAT*) **negative**
 OSTEOGENESIS IMPERFECTA TYPE VII (*CRTAP*) **negative**
 OSTEOGENESIS IMPERFECTA TYPE VIII (*P3H1*) **negative**
 OSTEOGENESIS IMPERFECTA TYPE XI (*FKBP10*) **negative**
 OSTEOGENESIS IMPERFECTA TYPE XIII (*BMP1*) **negative**
 OSTEOPECTOSIS, INFANTILE MALIGNANT, TCIRG1-RELATED (*TCIRG1*) **negative**
 OSTEOPECTOSIS, OSTM1-RELATED (*OSTM1*) **negative**

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

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P

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 (*SEP5C5*) **negative**
 PONTOCEREBELLAR HYPOPLASIA, VPS53-RELATED (*VPS53*) **negative**
 PRIMARY CILIARY DYSKINESIA, CCDC103-RELATED (*CCDC103*) **negative**
 PRIMARY CILIARY DYSKINESIA, CCDC39-RELATED (*CCDC39*) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAH11-RELATED (*DNAH11*) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAH5-RELATED (*DNAH5*) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAI1-RELATED (*DNAI1*) **negative**
 PRIMARY CILIARY DYSKINESIA, DNAI2-RELATED (*DNAI2*) **negative**
 PRIMARY CONGENITAL GLAUCOMA/PETERS ANOMALY (*CYP1B1*) **negative**
 PRIMARY HYPEROXALURIA, TYPE 1 (*AGXT*) **negative**
 PRIMARY HYPEROXALURIA, TYPE 2 (*GRHPR*) **negative**
 PRIMARY HYPEROXALURIA, TYPE 3 (*HOGA1*) **negative**
 PRIMARY MICROCEPHALY 1, AUTOSOMAL RECESSIVE (*MCPH1*) **negative**
 PROGRESSIVE EARLY-ONSET ENCEPHALOPATHY WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM (*TBCD*) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, ABCB4-RELATED (*ABCB4*) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 1 (*PFIC1*) (*ATP8B1*) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 2 (*ABCB11*) **negative**
 PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 4 (*PFIC4*) (*TJP2*) **negative**
 PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA (*CCN6*) **negative**
 PROLIDASE DEFICIENCY (*PEPD*) **negative**
 PROPIONIC ACIDEMIA, PCCA-RELATED (*PCCA*) **negative**
 PROPIONIC ACIDEMIA, PCCB-RELATED (*PCCB*) **negative**
 PSEUDOXANTHOMA ELASTICUM (*ABCC6*) **negative**
 PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (*PCD*) DEFICIENCY (*PCBD1*) **negative**
 PYCNODYSTOSIS (*CTS5K*) **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 IMMUNODYSPLASIA (*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), FOXP1-RELATED (*FOXP1*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), IKKB-RELATED (*IKKB*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), IL7R-RELATED (*IL7R*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), JAK3-RELATED (*JAK3*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), PTPRC-RELATED (*PTPRC*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY (SCID), RAG1-RELATED (*RAG1*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY, ADA-Related (*ADA*) **negative**
 SEVERE COMBINED IMMUNODEFICIENCY, TYPE ATHABASKAN (*DCLRE1C*) **negative**
 SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY (*DYNC2H1*) **negative**
 SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED (*SBDS*) **negative**
 SIALIDOSIS (*NEU1*) **negative**
 SJÖGREN-LARSSON SYNDROME (*ALDH3A2*) **negative**
 SMITH-LEMLI-OPITZ SYNDROME (*DHCR7*) **negative**
 SPASTIC PARAPLEGIA, TYPE 15 (*ZFYVE26*) **negative**

SPASTIC TETRAPLEGIA, THIN CORPUS CALLOSUM, AND PROGRESSIVE MICROCEPHALY (SPATCCM) (*SLC1A4*) **negative**
 SPG11-RELATED CONDITIONS (*SPG11*) **negative**
 SPINAL MUSCULAR ATROPHY (*SMN1*) **negative** SMN1: Two copies; g.27134T>G: absent; the absence of the g.27134T>G variant decreases the chance to be a silent (2+0) carrier.
 SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS TYPE 1 (*IGHMBP2*) **negative**
 SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 10 (*ANO10*) **negative**
 SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12 (*WWOX*) **negative**
 SPONDYLOCOSTAL DYSOSTOSIS 1 (*DLL3*) **negative**
 SPONDYLOTHORACIC DYSOSTOSIS, MESP2-Related (*MESP2*) **negative**
 STEEL SYNDROME (*COL27A1*) **negative**
 STERIOD-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**
 TRICHOHYDROSTROPHY 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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Z

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

Patient Information

Patient Name: [REDACTED]

Test Information

Ordering Physician: [REDACTED]



Clinic Information:

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date:

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, variants in exons 1-9 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.

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

For DDX11, only NM_030653.3:c.1763 - 1G > C variant will be analyzed and reported.

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

For OTOA, variants in exons 20 - 28 are not analyzed due to high sequence homology.

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

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

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 |

Patient Information

Patient Name: [REDACTED]

Test Information

Ordering Physician: [REDACTED]



Clinic Information: [REDACTED]

Date Of Birth: [REDACTED]

Case File ID: [REDACTED]

Report Date: [REDACTED]

Spinal Muscular Atrophy (SMN1)

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

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

Variant Classification

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

Negative Results

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

Additional Comments

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

PATIENT INFORMATION
7274, DONOR

| | |
|---------------|--------------|
| REPORT STATUS | Final |
|---------------|--------------|

Nichols Institute, Chantilly

DOB: [REDACTED] Age: [REDACTED]
 SEX: M

ORDERING PHYSICIAN

[REDACTED]

CLIENT INFORMATION

[REDACTED]

SPECIMEN INFORMATION

SPECIMEN: [REDACTED]
 REQUISITION: [REDACTED]
 LAB REF NO: [REDACTED]

ID: 7274-[REDACTED]

[REDACTED]

COLLECTED: 05/07/2024 00:00
 RECEIVED: 05/08/2024 19:13
 REPORTED: 05/14/2024 17:48

| Test Name | In Range | Out of Range | Reference Range | Lab |
|-----------------------------|----------|----------------|-------------------|-----|
| Hemoglobinopathy Evaluation | | | | AMD |
| Red Blood Cell Count | 5.04 | | 4.20-5.80 Mill/uL | |
| HEMOGLOBIN | 16.5 | | 13.2-17.1 g/dL | |
| Hematocrit | | | | |
| Hematocrit | | 51.0 H | 38.5-50.0 % | |
| MCV | | 101.2 H | 80.0-100.0 fL | |
| MCH | 32.7 | | 27.0-33.0 pg | |
| RDW | 12.5 | | 11.0-15.0 % | |
| Hemoglobin A | 97.1 | | >96.0 % | |
| Hemoglobin F | 0.0 | | <2.0 % | |
| Hemoglobin A2 (Quant) | 2.9 | | 2.2-3.2 % | |
| Interpretation | | | | |

NORMAL PATTERN

There is a normal pattern of hemoglobins and normal levels of Hb A2 and Hb F are present. No variant hemoglobins are observed. This is consistent with A/A phenotype. If iron deficiency coexists with a mild/silent beta thalassemia trait Hb A2 may be in the normal range. Rare variant hemoglobins have no separation from hemoglobin A by capillary zone electrophoresis (CZE) or high-performance liquid chromatography (HPLC). If clinically indicated, Thalassemia and Hemoglobinopathy Comprehensive (TC 17365) should be considered.

CBC (includes Differential and Platelets)
 CBC (includes Differential and Platelets)

AMD

| | | | |
|------------------------|------|----------------|-------------------|
| White Blood Cell Count | 7.4 | | 3.8-10.8 Thous/uL |
| Red Blood Cell Count | 5.04 | | 4.20-5.80 Mill/uL |
| HEMOGLOBIN | 16.5 | | 13.2-17.1 g/dL |
| Hematocrit | | 51.0 H | 38.5-50.0 % |
| MCV | | 101.2 H | 80.0-100.0 fL |
| MCH | 32.7 | | 27.0-33.0 pg |
| MCHC | 32.4 | | 32.0-36.0 g/dL |
| RDW | 12.5 | | 11.0-15.0 % |
| PLATELET COUNT | 253 | | 140-400 Thous/uL |
| MPV | 11.9 | | 7.5-12.5 fl |

PATIENT INFORMATION

7274, DONOR

REPORT STATUS **Final**

Nichols Institute, Chantilly

ORDERING PHYSICIAN

DOB: [REDACTED] Age: [REDACTED]

SEX: M

ID: 7274 [REDACTED]

COLLECTED: 05/07/2024 00:00

REPORTED: 05/14/2024 17:48

| Test Name | In Range | Out of Range | Reference Range | Lab |
|---|----------|--------------|--------------------|-----|
| CBC (includes Differential and Platelets) (Continued) | | | | |
| Absolute Neutrophils | 4877 | | 1500-7800 cells/uL | |
| Absolute Lymphocytes | 2065 | | 850-3900 cells/uL | |
| Absolute Monocytes | 318 | | 200-950 cells/uL | |
| Absolute Eosinophils | 81 | | 15-500 cells/uL | |
| Absolute Basophils | 59 | | 0-200 cells/uL | |
| Neutrophils | 65.9 | | % | |
| Lymphocytes | 27.9 | | % | |
| Monocytes | 4.30 | | % | |
| Eosinophils | 1.10 | | % | |
| Basophils | 0.80 | | % | |
| Nucleated RBC | 0.00 | | 0 /100 WBC | |

| | | | | |
|------------|------------|------------|------------|------------|
| [REDACTED] | [REDACTED] | [REDACTED] | [REDACTED] | [REDACTED] |
| [REDACTED] | [REDACTED] | [REDACTED] | [REDACTED] | [REDACTED] |
| [REDACTED] | [REDACTED] | [REDACTED] | [REDACTED] | [REDACTED] |

Chromosome Analysis, Blood

AMD

Chromosome Analysis, Blood

Chromosome Analysis, Blood

Order ID: [REDACTED]

Specimen Type: Blood

Clinical Indication: Semen donor

RESULT:
NORMAL MALE KARYOTYPE

INTERPRETATION:
Chromosome analysis revealed normal G-band patterns within the limits of standard cytogenetic analysis.

Please expect the results of any other concurrent study in a separate report.

NOMENCLATURE:
46,XY

ASSAY INFORMATION:

Method: G-Band (Digital Analysis:
MetaSystems/Ikaros)
Cells Counted: 20
Band Level: 550
Cells Analyzed: 5
Cells Karyotyped: 5

This test does not address genetic disorders that cannot be detected by standard cytogenetic methods or rare events such as low level mosaicism or subtle rearrangements.

Haiying Meng, M.D., Ph.D., FACMG, Technical Director, Cytogenetics and

PATIENT INFORMATION

7274, DONOR

REPORT STATUS **Final**

Nichols Institute, Chantilly

ORDERING PHYSICIAN

DOB: [REDACTED]

Age: [REDACTED]

SEX: M

ID: 7274-[REDACTED]

COLLECTED: 05/07/2024 00:00

REPORTED: 05/14/2024 17:48

| Test Name | In Range | Out of Range | Reference Range | Lab |
|-----------|----------|--------------|-----------------|-----|
|-----------|----------|--------------|-----------------|-----|

| | | | | |
|--|--|--|--|--|
| Chromosome Analysis, Blood (Continued) | | | | |
| Chromosome Analysis, Blood (Continued) | | | | |
| Genomics, 703-802-7156 | | | | |

Electronic Signature: 5/14/2024 5:04 PM

For additional information, please refer to <http://education.questdiagnostics.com/faq/chromsblood> (This link is being provided for informational/educational purposes only).

Performing Laboratory Information:

AMD Quest Diagnostics Nichols Institute 14225 Newbrook Drive Chantilly VA 20151 Laboratory Director: Patrick W Mason, MD PhD



Patient Information:

7274, Donor

DOB: [REDACTED]

Sex: M

MR#: 7274

Patient#: [REDACTED]

Accession:

[REDACTED]
Specimen Type: DNA
Collected: Not provided
Received Date: Aug 07,2025
Authorized Date: Aug 07,2025

Physician:

Wieloch, Shannon
GC: Wieloch, Shannon
Fairfax Cryobank
3015 Williams Drive #110
Fairfax, VA 22031
Phone: [REDACTED]
Fax:

Laboratory:

Fulgent Therapeutics LLC
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Amar Jariwala
Report Date: **Aug 20,2025**

Final Report

TEST PERFORMED

Single Gene - Gene

(1 Gene Panel: *PIEZO1*; gene sequencing with deletion and duplication analysis)

RESULTS:

No clinically significant sequence or copy-number variants were identified in the submitted specimen.

A negative result does not rule out the possibility of a genetic predisposition nor does it rule out any pathogenic mutations of the sort not queried by this test or in areas not reliably assessed by this test.

INTERPRETATION:

Notes and Recommendations:

- As requested, this report only includes variants classified as Pathogenic, Likely Pathogenic, or Risk Allele at the time of analysis. If detected, this report does not include variants classified as of uncertain significance.
- Gene specific notes and limitations may be present. See below.
- These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; <https://www.nsgc.org>)
- Guide to Interpreting Genomic Reports: A Genomics Toolkit (CSER Consortium; February 2017) (<https://www.genome.gov/For-Health-Professionals/Provider-Genomics-Education-Resources#hep>)

GENES TESTED:

Single Gene - Gene

1 genes tested (100.00% at >20x).

PIEZO1

Gene Specific Notes and Limitations

No gene specific limitations apply to the genes on the tested panel.

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 100.00% and 100.00% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications identified by NGS are confirmed by an orthogonal method (qPCR or MLPA), unless exceeding an internally specified and validated quality score, beyond which deletions and duplications are considered real without further confirmation. Bioinformatics: The FPLMv2.0 pipeline was used to analyze this specimen.

LIMITATIONS:

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to this individual's phenotype, and negative results do not rule out a genetic cause for the indication for testing. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (<https://www.genenames.org>) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is designed and validated for detection of germline variants only. It is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions (eg. trinucleotide or hexanucleotide repeat expansion). DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm for copy number variants, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which are two or more contiguous exons in size; single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

SIGNATURE:

A handwritten signature in black ink that reads "Harry Gao".

Dr. Harry Gao, DABMG, FACMG on 08/20/2025
Laboratory Director, Fulgent

DISCLAIMER:

This test was developed, performed, and its performance characteristics determined by **Fulgent Therapeutics LLC** (CAP# 8042697, CLIA# 05D2043189), 4399 Santa Anita Ave., El Monte, CA 91731. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at **(626) 350-0537** or info@fulgentgenetics.com. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.