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Genetic Testing Summary

Enclosed are the genetic testing results for

CB 595

No amount of genetic testing can guarantee that a child will not be affected with a genetic condition. Genetic testing can inform you of the likelihood of passing on the genetic conditions that are tested for, but it cannot eliminate the risk of passing on any genetic condition.

The genetic conditions Cryobio tests for are inherited in an autosomal recessive manner. This means that the child would have to inherit a genetic mutation from both the sperm source and the egg source to be affected with the condition. When both the sperm source and the egg source have undergone genetic carrier screening and the test results are negative, the risk of a child being affected with the conditions tested for is significantly reduced, but it cannot be completely eliminated.

All recipients should discuss both or their own risk for passing on genetic conditions and whether would benefit from genetic counseling and testing with their health care provider. Before using a donor that is a carrier for a specific recessive genetic condition or conditions, we strongly recommend that the recipient (or egg source, if different) consider genetic counseling and testing to determine if they are a carrier for the same genetic condition or conditions as the donor.

Screening and testing have changed dramatically over the years, and so the screening and testing done on each donor may vary depending on the testing that was in place when he was actively in Cryobio's donor program. Earlier donors may not have had as extensive testing as later donors. Screening and testing may change again in the future, so please review the results each time before ordering as both the testing done and the results may change.

Patient Information

Patient Name: CB 595

Date Of Birth: [REDACTED]/1999

Gender: Male

Ethnicity: Other

Patient ID: N/A

Medical Record #: N/A

Collection Kit: 36952899-2-C

Accession ID: N/A

Case File ID: 14057287

Test Information

Ordering Physician: MD David Prescott

Clinic Information: Cryobio Ohio

Phone: (614) 451-4375

Report Date: 10/09/2024

Sample Collected: 09/26/2024

Sample Received: 09/27/2024

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 557 out of 557 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.

CRYOBIO INTERNAL DATA ENTRY:
ENTERED BY: *CSB* **DATE:** 2-26-26
VERIFIED BY: *OK* **DATE:** 2-26-26

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Patient Information

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Date Of Birth: [REDACTED]/1999

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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**
ALKAPTONURIA (*HGD*) **negative**
ALPHA-1 ANTITRYPSIN DEFICIENCY (*SERPINA1*) **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**
ATRAFERRINEMIA (*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**
BERNARD-SOULIER SYNDROME, TYPE A1 (*GP1BA*) **negative**
BERNARD-SOULIER SYNDROME, TYPE C (*GP9*) **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**
CHOREOACANTHOCTOSIS (*VPS13A*) **negative**
CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (*CYBA*) **negative**
CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (*NCF2*) **negative**
CILIOPATHIES, RPGRIP1L-RELATED (*RPGRIP1L*) **negative**
CITRIN DEFICIENCY (*SLC25A13*) **negative**
CITRULLINEMIA, TYPE 1 (*ASS1*) **negative**
CLN10 DISEASE (*CTSD*) **negative**
COHEN SYNDROME (*VPS13B*) **negative**
COL11A2-RELATED CONDITIONS (*COL11A2*) **negative**
COMBINED MALONIC AND METHYLMALONIC ACIDURIA (*ACSF3*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (*GFM1*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (*TSM*) **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 (*MPL*) **negative**
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (*ALG6*) **negative**
CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (*SEC23B*) **negative**
CONGENITAL FINNISH NEPHROSIS (*NPHS1*) **negative**
CONGENITAL HYDROCEPHALUS 1 (*CCDC88C*) **negative**
CONGENITAL HYPERINSULINISM, KCNJ11-Related (*KCNJ11*) **negative**
CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (CIPA) (*NTRK1*) **negative**
CONGENITAL MYASTHENIC SYNDROME, CHAT-RELATED (*CHAT*) **negative**
CONGENITAL MYASTHENIC SYNDROME, CHRNE-RELATED (*CHRNE*) **negative**
CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED (*COLQ*) **negative**
CONGENITAL MYASTHENIC SYNDROME, DOK7-RELATED (*DOK7*) **negative**
CONGENITAL MYASTHENIC SYNDROME, RAPSN-RELATED (*RAPSN*) **negative**
CONGENITAL NEPHROTIC SYNDROME, PLCE1-RELATED (*PLCE1*) **negative**
CONGENITAL NEUTROPENIA, G6PC3-RELATED (*G6PC3*) **negative**
CONGENITAL NEUTROPENIA, HAX1-RELATED (*HAX1*) **negative**
CONGENITAL NEUTROPENIA, VPS45-RELATED (*VPS45*) **negative**
CONGENITAL SECRETORY CHLORIDE DIARRHEA 1 (*SLC26A3*) **negative**
CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS (*SLC4A11*) **negative**
CORTICOSTERONE METHYLOXIDASE DEFICIENCY (*CYP11B2*) **negative**
COSTEFF SYNDROME (3-METHYLGLUTACONIC ACIDURIA, TYPE 3) (*OPA3*) **negative**
CRB1-RELATED RETINAL DYSTROPHIES (*CRB1*) **negative**
CYSTIC FIBROSIS (*CFTR*) **negative**

Patient Information

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Test Information

Ordering Physician: MD David Prescott

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**C**

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

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, STXP2-RELATED (STXP2) **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 MEDITERRANEAN FEVER (MEFV) **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**
 HOMOCYSTEINURIA AND MEGALOBlastic ANEMIA TYPE CBLG (MTR) **negative**
 HOMOCYSTEINURIA DUE TO DEFICIENCY OF MTHFR (MTHFR) **negative**
 HOMOCYSTEINURIA, CBS-RELATED (CBS) **negative**
 HOMOCYSTEINURIA, Type cblE (MTRR) **negative**
 HYDROLETHALUS SYNDROME (HLYS1) **negative**
 HYPER-IGM IMMUNODEFICIENCY (CD40) **negative**
 HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA (HHH SYNDROME) (SLC25A15) **negative**
 HYPERPHOSPHATEMIC FAMILIAL TUMORAL CALCINOSIS, GALNT3-RELATED (GALNT3) **negative**
 HYPOMYELINATING LEUKODYSTROPHY 12 (VPS11) **negative**
 HYPOPHOSPHATASIA, ALPL-RELATED (ALPL) **negative**

I

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

J

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

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J
 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 AIP1 (AIP1) **negative**
 LEBER CONGENITAL AMAUROSIS TYPE GUCY2D (GUCY2D) **negative**
 LEBER CONGENITAL AMAUROSIS TYPE TULP1 (TULP1) **negative**
 LEBER CONGENITAL AMAUROSIS, IQCB1-RELATED/SENIOR-LOKEN SYNDROME 5 (IQCB1) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE CEP290 (CEP290) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE LCA5 (LCA5) **negative**
 LEBER CONGENITAL AMAUROSIS, TYPE RDH12 (RDH12) **negative**
 LEIGH SYNDROME, FRENCH-CANADIAN TYPE (LRPPRC) **negative**
 LETHAL CONGENITAL CONTRACTURE SYNDROME 1 (GLE1) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER (EIF2B5) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B1-RELATED (EIF2B1) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B2-RELATED (EIF2B2) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B3-RELATED (EIF2B3) **negative**
 LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B4-RELATED (EIF2B4) **negative**
 LIG4 SYNDROME (LIG4) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 8 (TRIM32) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2A (CAPN3) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2B (DYSF) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2C (SGCG) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2D (SGCA) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2E (SGCB) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2F (SGCD) **negative**
 LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2I (FKRP) **negative**
 LIPOAMIDE DEHYDROGENASE DEFICIENCY (DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY) (DLD) **negative**
 LIPOID ADRENAL HYPERPLASIA (STAR) **negative**
 LIPOPROTEIN LIPASE DEFICIENCY (LPL) **negative**
 LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (HADHA) **negative**
 LRAT-RELATED CONDITIONS (LRAT) **negative**
 LUNG DISEASE, IMMUNODEFICIENCY, AND CHROMOSOME BREAKAGE SYNDROME (LICS) (NSMCE3) **negative**
 LYSINURIC PROTEIN INTOLERANCE (SLC7A7) **negative**

M
 MALONYL-COA DECARBOXYLASE DEFICIENCY (MLYCD) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 1A (BCKDHA) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 1B (BCKDHB) **negative**
 MAPLE SYRUP URINE DISEASE, TYPE 2 (DBT) **negative**
 MCKUSICK-KAUFMAN SYNDROME (MKKS) **negative**
 MECKEL SYNDROME 7/NEPHRONOPHTHISIS 3 (NPHP3) **negative**
 MECKEL-GRUBER SYNDROME, TYPE 1 (MKS1) **negative**
 MECR-RELATED NEUROLOGIC DISORDER (MECR) **negative**
 MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (ACADM) **negative**
 MEDNIK SYNDROME (AP1S1) **negative**
 MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS (MLC1) **negative**
 MEROSIN-DEFICIENT MUSCULAR DYSTROPHY (LAMA2) **negative**
 METABOLIC ENCEPHALOPATHY AND ARRHYTHMIAS, TANGO2-RELATED (TANGO2) **negative**
 METACHROMATIC LEUKODYSTROPHY, ARSA-RELATED (ARSA) **negative**
 METACHROMATIC LEUKODYSTROPHY, PSAP-RELATED (PSAP) **negative**
 METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CBLF (LMBRD1) **negative**
 METHYLMALONIC ACIDEMIA, MCEE-RELATED (MCEE) **negative**
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CBLC (MMACHC) **negative**
 METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CblD (MMADHC) **negative**
 METHYLMALONIC ACIDURIA, MMAA-RELATED (MMAA) **negative**
 METHYLMALONIC ACIDURIA, MMAB-RELATED (MMAB) **negative**
 METHYLMALONIC ACIDURIA, TYPE MUT (0) (MUT) **negative**
 MEVALONIC KINASE DEFICIENCY (MVK) **negative**
 MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II (PCNT) **negative**
 MICROPHthalmia / ANOPHTHALMIA, VSX2-RELATED (VSX2) **negative**
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, ACAD9-RELATED (ACAD9) **negative**
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFAF5-RELATED (NDUFAF5) **negative**
 MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFS6-RELATED (NDUFS6) **negative**
 MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 1 (NDUFS4) **negative**

MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 10 (NDUFA2) **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, PJKV-RELATED (PJKV) **negative**
 NONSYNDROMIC HEARING LOSS, SYNE4-RELATED (SYNE4) **negative**
 NONSYNDROMIC HEARING LOSS, TMC1-RELATED (TMC1) **negative**
 NONSYNDROMIC HEARING LOSS, TMPRSS3-RELATED (TMPRSS3) **negative**
 NONSYNDROMIC INTELLECTUAL DISABILITY (CC2D1A) **negative**
 NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (SAMD9) **negative**

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

P
 PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION (PANK2) **negative**

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**P**

PAPILLON LEFÈVRE SYNDROME (CTSC) **negative**
 PARKINSON DISEASE 15 (FBX07) **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**
 PSEUDOCHELINESTERASE DEFICIENCY (BCHIE) **negative**
 PSEUDOXANTHOMA ELASTICUM (ABCC6) **negative**
 PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (PCD) DEFICIENCY (PCBD1) **negative**
 PYCNODYSTOSIS (CTSK) **negative**
 PYRIDOXAL 5'-PHOSPHATE-DEPENDENT EPILEPSY (PNPO) **negative**
 PYRIDOXINE-DEPENDENT EPILEPSY (ALDH7A1) **negative**
 PYRUVATE CARBOXYLASE DEFICIENCY (PC) **negative**
 PYRUVATE DEHYDROGENASE DEFICIENCY, PDHB-RELATED (PDHB) **negative**

R

REFSUM DISEASE, PHYH-RELATED (PHYH) **negative**
 RENAL TUBULAR ACIDOSIS AND DEAFNESS, ATP6V1B1-RELATED (ATP6V1B1) **negative**
 RENAL TUBULAR ACIDOSIS, PROXIMAL, WITH OCULAR ABNORMALITIES AND MENTAL RETARDATION (SLC4A4) **negative**
 RETINITIS PIGMENTOSA 25 (EYS) **negative**
 RETINITIS PIGMENTOSA 26 (CERKL) **negative**
 RETINITIS PIGMENTOSA 28 (FAM161A) **negative**
 RETINITIS PIGMENTOSA 36 (PRCD) **negative**
 RETINITIS PIGMENTOSA 59 (DHDDS) **negative**
 RETINITIS PIGMENTOSA 62 (MAK) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1 (PEX7) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2 (GNPAT) **negative**
 RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3 (AGP5) **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 IMMUNOOSSOUS 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 POLYDACTYL (DYNC2H1) **negative**
 SHWACHMAN-DIAMOND SYNDROME, SBDS-RELATED (SBDS) **negative**
 SIALIDOSIS (NEU1) **negative**
 SJÖGREN-LARSSON SYNDROME (ALDH3A2) **negative**
 SMITH-LEMMLI-OPITZ SYNDROME (DCHR7) **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**
 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**
 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**

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

Patient Information

Patient Name: CB 595

Date Of Birth: [REDACTED]/1999

Case File ID: 14057287

Test Information

Ordering Physician: MD David Prescott

Clinic Information: Cryobio Ohio

Report Date: 10/09/2024

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

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

For OTOA, 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: CB 595

Date Of Birth: [REDACTED]/1999

Case File ID: 14057287

Test Information

Ordering Physician: MD David Prescott

Clinic Information: Cryobio Ohio

Report Date: 10/09/2024

**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.

Client/Sending Facility:
Cryobio

4845 Knightsbridge Blvd., Ste 200
Columbus, OH 43214
Ph: (614)451-4375
OHB-12

LCLS Specimen Number: 270-488-8546-0

Patient Name: CB, 595

Date of Birth: [REDACTED] 1999

Gender: M

Patient ID:

Lab Number: YU24-102175 L

Indications: NOT GIVEN

Account Number: 34334785

Ordering Physician:

Specimen Type: BLOOD

Client Reference:

Date Collected: 09/26/2024

Date Received: 09/30/2024

Date Reported: 10/09/2024

Test: Chromosome, Blood, Routine

Cells Counted: 20

Cells Analyzed: 20

Cells Karyotyped: 2

Band Resolution: 500

CYTOGENETIC RESULT: 46,XY

INTERPRETATION: NORMAL MALE KARYOTYPE

Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTG banding pattern in all cells observed.

This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.

Technical Component- Partial chromosome analysis performed by LabCorp, CLIA 45D0674994, 1522 Flanders Field Lane, Sugarland, TX 77498. Laboratory Director, Venkateswara R Potluri PhD.

Technical Component- Partial chromosome analysis performed by LabCorp, CLIA 45D0674994. 296 Hawthorne Drive Madison, Mississippi 39110. Laboratory Director, Venkateswara R Potluri PhD.

Technical Component-Processing performed at 1904 TW Alexander Dr, Research Triangle Park, NC 27709, Labcorp CLIA 34D1008914. Medical Director, Anjen Chenn, M.D., Ph.D.

CRYOBIO INTERNAL DATA ENTRY:
ENTERED BY: *CB* **DATE:** 2-26-26
VERIFIED BY: *OA* **DATE:** 2-26-26

LCLS Specimen Number: 270-488-8546-0

Patient Name: **CB, 595**

Date of Birth: [REDACTED] 1999

Gender: M

Patient ID:

Lab Number: YU24-102175 L

Account Number: 34334785

Ordering Physician:

Specimen Type: **BLOOD**

Client Reference:

Date Collected: 09/26/2024

Date Received: 09/30/2024



Client/Sending Facility:
Cryobio

4845 Knightsbridge Blvd., Ste 200
Columbus, OH 43214
Ph: (614)451-4375
OHB-12

LCLS Specimen Number: 270-488-8546-0

Patient Name: **CB, 595**

Date of Birth: [REDACTED]/1999

Gender: M

Patient ID:

Lab Number: YU24-102175 L

Account Number: 34334785

Ordering Physician:

Specimen Type: **BLOOD**

Client Reference:

Date Collected: 09/26/2024

Date Received: 09/30/2024



Anh Vu, PhD, FACMG

Anjen Chenn, M.D., Ph.D.
Medical Director

Technical component performed by Laboratory Corporation of America Holdings,
1904 TW Alexander Drive , RTP , NC , 27709-0153 (800) 345-4363

Professional Component performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr, Research Triangle Park, NC 27709. Medical Director, Anjen Chenn, M.D., Ph.D.
Integrated Genetics is a brand used by Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

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If you have received this document in error, please call 800-533-0567.



Date Collected: 09/26/2024 Date Received: 09/26/2024 Date Reported: 10/10/2024 Fasting: Not Given

Ordered Items: Chromosome, Blood, Routine; Count 15-20 cells, 2 Karyotype; Chromosome Blood Routine 88230

Date Collected: 09/26/2024

Chromosome, Blood, Routine

Test	Current Result and Flag	Previous Result and Date	Units	Reference Interval
Specimen Type ⁰¹	Comment: BLOOD			
Cells Counted ⁰¹	20			
Cells Analyzed ⁰¹	20			
Cells Karyotyped ⁰¹	2			
GTG Band Resolution Achieved ⁰¹	500			
Cytogenetic Result ⁰¹	Comment: 46, XY			
Interpretation ⁰¹	<p>Comment:</p> <p>NORMAL MALE KARYOTYPE</p> <p>Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTG banding pattern in all cells observed.</p> <p>This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.</p> <p>Technical Component- Partial chromosome analysis performed by LabCorp, CLIA 45D0674994, 1522 Flanders Field Lane, Sugarland, TX 77498. Laboratory Director, Venkateswara R Potluri PhD.</p> <p>Technical Component- Partial chromosome analysis performed by LabCorp, CLIA 45D0674994. 296 Hawthorne Drive Madison, Mississippi 39110. Laboratory Director, Venkateswara R Potluri PhD.</p> <p>Technical Component-Processing performed at 1904 TW Alexander Dr, Research Triangle Park, NC 27709, Labcorp CLIA 34D1008914. Medical Director, Anjen Chenn, M.D., Ph.D.</p>			
Director Review: ⁰¹	Comment: Anh Vu, PhD, FACMG			
PDF				

Disclaimer

The Previous Result is listed for the most recent test performed by Labcorp in the past 5 years where there is sufficient patient demographic data to match the result to the patient. Results from certain tests are excluded from the Previous Result display.

Icon Legend

▲ Out of Reference Range ■ Critical or Alert

Performing Labs

01: YU - Labcorp RTP, 1904 TW Alexander Drive Ste C, RTP, NC 27709-0153 Dir: Anjen Chenn, MD PhD
For Inquiries, the physician may contact Branch: 800-321-3862 Lab: 800-282-7300

CRYOBIO INTERNAL DATA ENTRY:
ENTERED BY: *ESD* DATE: 2-26-26
VERIFIED BY: *OK* DATE: 2-26-26

Cb, 595

Patient ID:
Specimen ID: 270-488-8546-0

DOB: [REDACTED] 1999

Age: 25
Sex: Male

Patient Report

Account Number: 34334785
Ordering Physician:



Patient Details
Cb, 595

Phone:
Date of Birth: [REDACTED] 1999
Age: 25
Sex: Male
Patient ID:
Alternate Patient ID:

Physician Details

Cryobio
4845 Knightsbridge Blvd., Ste 200,
Columbus, OH, 43214

Phone: 614-451-4375
Account Number: 34334785
Physician ID:
NPI:

Specimen Details

Specimen ID: 270-488-8546-0
Control ID: A9S34334785
Alternate Control Number:
Date Collected: 09/26/2024 1530 Local
Date Received: 09/26/2024 0000 ET
Date Entered: 09/26/2024 2139 ET
Date Reported: 10/10/2024 0811 ET

Cb, 595

Patient ID:
Specimen ID: 215-488-8510-0

DOB: [REDACTED]/1999

Age: 24
Sex: Male

Patient Report

Account Number: 34334785
Ordering Physician:



Date Collected: 08/02/2024

Date Received: 08/02/2024

Date Reported: 08/05/2024

Fasting: No

Ordered Items: CBC With Differential/Platelet; Hgb Fractionation Cascade

Date Collected: 08/02/2024

ENTERED: CF 8-29-24
VERIFIED: OH 8-29-24

CBC With Differential/Platelet

Test	Current Result and Flag	Previous Result and Date	Units	Reference Interval
WBC ⁰¹	7.6		x10E3/uL	3.4-10.8
RBC ⁰¹	4.98		x10E6/uL	4.14-5.80
Hemoglobin ⁰¹	15.0		g/dL	13.0-17.7
Hematocrit ⁰¹	47.0		%	37.5-51.0
MCV ⁰¹	94		fL	79-97
MCH ⁰¹	30.1		pg	26.6-33.0
MCHC ⁰¹	31.9		g/dL	31.5-35.7
RDW ⁰¹	12.6		%	11.6-15.4
Platelets ⁰¹	244		x10E3/uL	150-450
Neutrophils ⁰¹	42		%	Not Estab.
Lymphs ⁰¹	38		%	Not Estab.
Monocytes ⁰¹	7		%	Not Estab.
Eos ⁰¹	12		%	Not Estab.
Basos ⁰¹	1		%	Not Estab.
Neutrophils (Absolute) ⁰¹	3.3		x10E3/uL	1.4-7.0
Lymphs (Absolute) ⁰¹	2.9		x10E3/uL	0.7-3.1
Monocytes(Absolute) ⁰¹	0.5		x10E3/uL	0.1-0.9
▲ Eos (Absolute) ⁰¹	0.9 High		x10E3/uL	0.0-0.4
Baso (Absolute) ⁰¹	0.1		x10E3/uL	0.0-0.2
Immature Granulocytes ⁰¹	0		%	Not Estab.
Immature Grans (Abs) ⁰¹	0.0		x10E3/uL	0.0-0.1

Hgb Fractionation Cascade

Test	Current Result and Flag	Previous Result and Date	Units	Reference Interval
Hgb Fractionation by CE: ⁰¹				
Hgb F ⁰¹	0.0		%	0.0-2.0
Hgb A ⁰¹	97.4		%	96.4-98.8
Hgb A2 ⁰¹	2.6		%	1.8-3.2
Hgb S ⁰¹	0.0		%	0.0

Interpretation:⁰¹

Normal hemoglobin present; no hemoglobin variant or beta thalassemia identified.

Note: Alpha thalassemia may not be detected by the Hgb Fractionation Cascade panel. If alpha thalassemia is suspected, Labcorp offers Alpha-Thalassemia DNA Analysis (#511172).

X David Prescott, M.D.
Medical Director Review

8/16/2024
Date

labcorp

Date Created and Stored 08/05/24 1515 ET Final Report Page 1 of 2

Cb, 595

Patient ID:
Specimen ID: 215-488-8510-0

DOB: [REDACTED]/1999

Age: 24
Sex: Male

Patient Report

Account Number: 34334785
Ordering Physician:



Disclaimer

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Icon Legend

▲ Out of Reference Range ■ Critical or Alert

Performing Labs

01: CB - Labcorp Dublin, 6370 Wilcox Road, Dublin, OH 43016-1269 Dir: Vincent Ricchiuti, PhD
For Inquiries, the physician may contact Branch: 800-321-3862 Lab: 800-282-7300

Patient Details

Cb, 595

Phone:
Date of Birth: [REDACTED]/1999
Age: 24
Sex: Male
Patient ID:
Alternate Patient ID:

Physician Details

Cryobio
4845 Knightsbridge Blvd., Ste 200,
Columbus, OH, 43214

Phone: 614-451-4375
Account Number: 34334785
Physician ID:
NPI:

Specimen Details

Specimen ID: 215-488-8510-0
Control ID: A4U34334785
Alternate Control Number:
Date Collected: 08/02/2024 0830 Local
Date Received: 08/02/2024 0000 ET
Date Entered: 08/02/2024 2128 ET
Date Reported: 08/05/2024 1506 ET