Patient Information	
Patient Name:	Elizabeth Bennett
Date Of Birth:	01/12/1996
Gender:	Female
Ethnicity:	Sephardic Jewish
Patient ID:	N/A
Medical Record #:	N/A
Collection Kit:	7565288-2-C
Accession ID:	N/A
Case File ID:	3113050

Test Information	
Ordering Physician:	N/A
Clinic Information:	Natera, Inc.
Phone:	555-555-5555
Report Date:	01/19/2021
Sample Collected:	12/28/2020
Sample Received:	12/29/2020
Sample Type:	Blood

horizon™ natera carrier screen

CARRIER SCREENING REPORT

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

ORDER SELECTED: The Horizon **421** panel was ordered for this patient.

FINAL RESULTS SUMMARY:

CARRIER for Congenital Myasthenic Syndrome, COLQ-Related

Positive for the pathogenic variant exon 1 deletion in the COLQ gene. If this individual's partner is a carrier for CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED, their chance to have a child with this condition is 1 in 4 (25%). Carrier screening for this individual's partner is suggested.

Negative for 420 out of 421 diseases

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

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

RECOMMENDATIONS

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

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Laboratory Director, Natera

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Patient Information		Test Information	
Patient Name:	Elizabeth Bennett	Ordering Physician:	N/A
		Clinic Information:	Natera, Inc.
Date Of Birth:	01/12/1996		
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CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED

Understanding Your Horizon Carrier Screen Results: CONGENITAL MYASTHENIC SYNDROME, COLQ-RELATED

What is Congenital Myasthenic Syndrome, COLQ-Related?

Congenital Myasthenic Syndrome, COLQ-Related (also called CMS5) is an inherited disorder that affects the muscles. Muscle weakness (myasthenia) typically begins shortly after birth along with poor muscle tone (hypotonia) and episodes of apnea (periodic stops in breathing). In some cases, symptoms may not begin until later in life. Affected infants and children often have feeding and swallowing problems, developmental delays, and breathing problems that include episodes of apnea that get worse during illness. Muscle weakness may worsen with exercise. Speech problems may occur due to facial muscle weakness. The degree of muscle weakness varies among individuals affected with Congenital Myasthenic Syndrome, COLQ-Related, but usually remains stable and does not worsen with age. Currently, there is no cure for this disorder and treatment is based on symptoms. Clinical trials involving potential new treatments for this condition may be available (see www.clinicaltrials.gov).

What causes Congenital Myasthenic Syndrome, COLQ-Related?

The majority of cases of Congenital Myasthenic Syndrome, COLQ-Related are caused by a change, or mutation, in both copies of the COLQ gene pair. These mutations cause the genes to not work properly or not work at all. When both copies of the COLQ gene do not work correctly, it leads to the symptoms described above. Most cases of Congenital Myasthenic Syndrome, COLQ-Related are inherited in an autosomal recessive manner. This means that, in most cases, both parents must be carriers of a mutation in one copy of the COLQ gene to have a child with this type of Congenital Myasthenic Syndrome. Most people who are carriers for Congenital Myasthenic Syndrome, COLQ-Related are healthy and do not have symptoms, nor do they have this disorder themselves. Usually a child inherits two copies of each gene, one copy from the mother and one copy from the father. If the mother and father are both carriers for Congenital Myasthenic Syndrome, COLQ-Related, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their COLQ gene mutations to the child, who will then have the disorder. Individuals found to carry more than one mutation for Congenital Myasthenic Syndrome, COLQ-Related should discuss their risk for having an affected child, and any potential effects to their own health, with their health care provider. There are a number of other forms of Congenital Myasthenic Syndrome, each caused by mutations in different genes. A person who is a carrier for Congenital Myasthenic Syndrome, COLQ-Related is not likely to have an increased risk of having children with these other forms.

What can I do next?

You may wish to speak with a local genetic counselor about your carrier test results. A genetic counselor in your area can be located on the National Society of Genetic Counselors website (www.nsgc.org). Your siblings and other relatives are at increased risk to also have this mutation. You are encouraged to inform your family members of your test results as they may wish to consider being tested themselves. If you are pregnant, your partner can have carrier screening for Congenital Myasthenic Syndrome, COLQ-Related ordered by a health care professional. If your partner is not found to be a carrier for Congenital Myasthenic Syndrome, COLQ-Related, your risk of having an affected child is greatly reduced. Couples at risk of having a baby with Congenital Myasthenic Syndrome, COLQ-Related can opt to have prenatal diagnosis done through chorionic villus sampling (CVS) or amniocentesis during pregnancy or can choose to have the baby tested after birth for this condition. If you are not yet pregnant, your partner can have carrier screening for Congenital Myasthenic Syndrome, COLQ-Related ordered by a health care professional. If your partner is found to be a carrier for this disorder, you have several reproductive options to consider:

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

What resources are available?

- Genetics Home Reference: http://ghr.nlm.nih.gov/condition/congenital-myasthenic-syndrome
- GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1168/
- Prenatal diagnosis done through CVS: http://www.marchofdimes.org/chorionic-villus-sampling.aspx
- Prenatal diagnosis done through Amniocentesis: http://www.marchofdimes.org/amniocentesis.aspx
- PGD with IVF: http://www.natera.com/spectrum

Patient Information
Patient Name: Elizabeth Bennett

Date Of Birth: 01/12/1996
Case File ID: 3113050

Test Information
Ordering Physician: N/A

Clinic Information: Natera, Inc.

Report Date: 01/19/2021

horizon™ natera carrier screen

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

C
CEREBROOCULOFACIOSKELETAL SYNDROME 1 (COFS1) (*ERCC6*) **negative**
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**

6
6-PYRUVOYL-TETRAHYDROPTERIN SYNTHASE (PTPS) DEFICIENCY (*PTS*) **negative**

A
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-GOUTIÃˆRES SYNDROME, RNASEH2A-RELATED (*RNASEH2A*) **negative**
AICARDI-GOUTIÃˆRES SYNDROME, RNASEH2B-RELATED (*RNASEH2B*) **negative**
AICARDI-GOUTIÃˆRES SYNDROME, RNASEH2C-RELATED (*RNASEH2C*) **negative**
AICARDI-GOUTIÈRES SYNDROME (*SAMHD1*) **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**
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**
AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (*SLC35A3*) **negative**
AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (*AIRE*) **negative**
AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (*SACS*) **negative**

B
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, 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**
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-UREIDOPROPIONASE DEFICIENCY (*UPB1*) **negative**
BILATERAL FRONTOPARIETAL POLYMICROGYRIA (*GPR56*) **negative**
BIOTINIDASE DEFICIENCY (*BTD*) **negative**
BLOOM SYNDROME (*BLM*) **negative**

C
CANAVAN DISEASE (*ASPA*) **negative**
CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (*CPS1*) **negative**
CARNITINE DEFICIENCY (*SLC22A5*) **negative**
CARNITINE PALMITOYLTRANSFERASE I A DEFICIENCY (*CPT1A*) **negative**
CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (*CPT2*) **negative**
CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (*SLC25A20*) **negative**
CARPENTER SYNDROME (*RAB23*) **negative**
CARTILAGE-HAIR HYPOPLASIA (*RMRP*) **negative**
CEREBROTENDINOUS XANTHOMATOSIS (*CYP27A1*) **negative**
CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (*NDRG1*) **negative**
CHEDIAK-HIGASHI SYDROME (*LYST*) **negative**
CHOREOACANTHOCYTOSIS (*VPS13A*) **negative**
CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (*CYBA*) **negative**
CILIOPATHIES, RPGRIP1L-RELATED (*RPGRIP1L*) **negative**
CITRIN DEFICIENCY (*SLC25A13*) **negative**
CITRULLINEMIA, TYPE 1 (*ASS1*) **negative**
CLN10 DISEASE (*CTSD*) **negative**
COHEN SYNDROME (*VPS13B*) **negative**
COMBINED MALONIC AND METHYLMALONIC ACIDURIA (*ACSF3*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (*GFM1*) **negative**
COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (*TSFM*) **negative**
COMBINED PITUITARY HORMONE DEFICIENCY-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 AMEGAKARYOCYTIC THROMBOCYTOPENIA (*MPL*) **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 FINNISH NEPHROSIS (*NPHS1*) **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*) **see first page**
CONGENITAL MYASTHENIC SYNDROME, DOK7-RELATED (*DOK7*) **negative**
CONGENITAL MYASTHENIC SYNDROME, RAPSN-RELATED (*RAPSN*) **negative**
CONGENITAL NEPHROTIC SYNDROME, PLCE1-RELATED (*PLCE1*) **negative**
CONGENITAL NEUTROPENIA, HAX1-RELATED (*HAX1*) **negative**
CONGENITAL NEUTROPENIA, VPS45-RELATED (*VPS45*) **negative**
CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS (*SLC4A11*) **negative**
CORTICOSTERONE METHYLOXIDASE DEFICIENCY (*CYP11B2*) **negative**
COSTEFF SYNDROME (3-METHYLGLUTACONIC ACIDURIA, TYPE 3) (*OPA3*) **negative**
CRB1-RELATED RETINAL DYSTROPHIES (*CRB1*) **negative**
CYSTIC FIBROSIS (*CFTR*) **negative**
CYSTINOSIS (*CTNS*) **negative**
CYTOCHROME C OXIDASE DEFICIENCY, PET100-RELATED (*PET100*) **negative**

D
D-BIFUNCTIONAL PROTEIN DEFICIENCY (*HSD17B4*) **negative**
DEAFNESS, AUTOSOMAL RECESSIVE 77 (*LOXHD1*) **negative**
DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY (*DPYD*) **negative**
DYSKERATOSIS CONGENITA, RTEL1-RELATED (*RTEL1*) **negative**
DYSTROPHIC EPIDERMOLYSIS BULLOSA, COL7A1-Related (*COL7A1*) **negative**

E
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**

E
EPIPHYSEAL DYSPLASIA, MULTIPLE, 7 / DESBUQUOIS DYSPLASIA 1 (*CANT1*) **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 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 L (*FANCL*) **negative**
FARBER LIPOGRANULOMATOSIS (*ASAH1*) **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**
GITELMAN SYNDROME (*SLC12A3*) **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**
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 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**
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 SPASTIC PARAPARESIS, TYPE 49 (*TECPR2*) **negative**
HERMANSKY-PUDLAK SYNDROME, AP3B1-RELATED (*AP3B1*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED (*HPS1*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS3-RELATED (*HPS3*) **negative**
HERMANSKY-PUDLAK SYNDROME, HPS4-RELATED (*HPS4*) **negative**
HOLOCARBOXYLASE SYNTHETASE DEFICIENCY (*HLCS*) **negative**
HOMOCYSTINURIA DUE TO DEFICIENCY OF MTHFR (*MTHFR*) **negative**
HOMOCYSTINURIA, CBS-RELATED (*CBS*) **negative**
HOMOCYSTINURIA, Type cblE (*MTRR*) **negative**
HYDROLETHALUS SYNDROME (*HYLS1*) **negative**
HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA (HHH SYNDROME) (*SLC25A15*) **negative**
HYPERPHOSPHATEMIC FAMILIAL TUMORAL CALCINOSIS, GALNT3-RELATED (*GALNT3*) **negative**
HYPOPHOSPHATASIA, ALPL-RELATED (*ALPL*) **negative**

I
INCLUSION BODY MYOPATHY 2 (*GNE*) **negative**
INFANTILE CEREBRAL AND CEREBELLAR ATROPHY (*MED17*) **negative**
INFANTILE NEPHRONOPHTHISIS (*INVS*) **negative**
INFANTILE NEUROAXONAL DYSTROPHY (*PLA2G6*) **negative**
ISOVALERIC ACIDEMIA (*IVD*) **negative**

J
JOHANSON-BLIZZARD SYNDROME (*UBR1*) **negative**
JOUBERT SYNDROME 2 / MECKEL SYNDROME 2 (*TMEM216*) **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, LAMB3-RELATED (*LAMB3*) **negative**
JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMC2-RELATED (*LAMC2*) **negative**
JUNCTIONAL EPIDERMOLYSIS BULLOSA/LARYNGOONYCHOCUTANEOUS SYNDROME, LAMA3-RELATED (*LAMA3*) **negative**

K
KRABBE DISEASE (*GALC*) **negative**

L
LAMELLAR ICHTHYOSIS, TYPE 1 (*TGM1*) **negative**
LEBER CONGENITAL AMAUROSIS 2 (*RPE65*) **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**
LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2 E (*SGCB*) **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 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**
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 SYDROME (*MKKS*) **negative**
MECKEL SYNDROME 7/ NEPHRONOPHTHISIS 3 (*NPHP3*) **negative**
MECKEL-GRUBER SYNDROME, TYPE 1 (*MKS1*) **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 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**
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 17 (*NDUFAF6*) **negative**
MITOCHONDRIAL MYOPATHY AND SIDEROBLASTIC ANEMIA (MLASA1) (*PUS1*) **negative**
MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (*HADHB*) **negative**

M
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 B / GM1 GANGLIOSIDOSIS (*GLB1*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE IVA (MORQUIO SYNDOME) (*GALNS*) **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**
MYONEUROGASTROINTESTINAL ENCEPHALOPATHY (MNGIE) (*TYMP*) **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**
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**

O
ODONTO-ONYCHO-DERMAL DYSPLASIA / SCHOPF-SCHULZ-PASSARGE SYNDROME (*WNT10A*) **negative**
OMENN SYNDROME, RAG2-RELATED (*RAG2*) **negative**
ORNITHINE AMINOTRANSFERASE DEFICIENCY (*OAT*) **negative**
OSTEOPETROSIS, INFANTILE MALIGNANT, TCIRG1-RELATED (*TCIRG1*) **negative**

P
PENDRED SYNDROME (*SLC26A4*) **negative**
PERLMAN SYNDROME (*DIS3L2*) **negative**
PHENYLKETONURIA (*PAH*) **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, 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**
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**
PROLIDASE DEFICIENCY (*PEPD*) **negative**
PROPIONIC ACIDEMIA, PCCA-RELATED (*PCCA*) **negative**
PROPIONIC ACIDEMIA, PCCB-RELATED (*PCCB*) **negative**
PSEUDOCHOLINESTERASE DEFICIENCY (*BCHE*) **negative**
PSEUDOXANTHOMA ELASTICUM (*ABCC6*) **negative**
PYCNODYSOSTOSIS (*CTSK*) **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 59 (*DHDDS*) **negative**
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1 (*PEX7*) **negative**
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2 (*GNPAT*) **negative**
RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3 (*AGPS*) **negative**
ROBERTS SYNDROME (*ESCO2*) **negative**

S
SALLA DISEASE (*SLC17A5*) **negative**
SANDHOFF DISEASE (*HEXB*) **negative**
SCHIMKE IMMUNOOSSEOUS DYSPLASIA (*SMARCAL1*) **negative**
SEGAWA SYNDROME, TH-RELATED (*TH*) **negative**
SENIOR-LOKEN SYNDROME 4 / NEPHRONOPHTHISIS 4 (*NPHP4*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY (SCID), RAG1-RELATED (*RAG1*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY, ADA-Related (*ADA*) **negative**
SEVERE COMBINED IMMUNODEFICIENCY, TYPE ATHABASKAN (*DCLRE1C*) **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**
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.**
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12 (*WWOX*) **negative**
SPONDYLOTHORACIC DYSOSTOSIS, MESP2-Related (*MESP2*) **negative**
STEEL SYNDROME (*COL27A1*) **negative**
STEROID-RESISTANT NEPHROTIC SYNDROME (*NPHS2*) **negative**
STUVE-WIEDEMANN SYNDROME (*LIFR*) **negative**

T
TAY-SACHS DISEASE (*HEXA*) **negative**
TRICHOHEPATOENTERIC SYNDROME, TTC37-RELATED (*TTC37*) **negative**
TRICHOTHIODYSTROPHY 1 / XERODERMA PIGMENTOSUM, GROUP D (*ERCC2*) **negative**
TRIPLE A SYNDROME (*AAAS*) **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**
VITAMIN D-DEPENDENT RICKETS, TYPE 1A (*CYP27B1*) **negative**

W
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**
WERNER SYNDROME (*WRN*) **negative**
WILSON DISEASE (*ATP7B*) **negative**
WOLCOTT-RALLISON SYNDROME (*EIF2AK3*) **negative**
WOLMAN DISEASE (*LIPA*) **negative**

X
XERODERMA PIGMENTOSUM, GROUP A (*XPA*) **negative**
XERODERMA PIGMENTOSUM, GROUP C (*XPC*) **negative**

Z
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**

X-Linked

A
ADRENAL HYPOPLASIA CONGENITA, X-LINKED (*NR0B1*) **negative**
ADRENOLEUKODYSTROPHY, X-LINKED (*ABCD1*) **negative**
AGAMMAGLOBULINEMIA, X-LINKED (*BTK*) **negative**
ALPHA-THALASSEMIA INTELLECTUAL DISABILITY SYNDROME (*ATRX*) **negative**
ALPORT SYNDROME, X-LINKED (*COL4A5*) **negative**
ARTS SYNDROME (*PRPS1*) **negative**

B
BARTH SYNDROME (*TAZ*) **negative**

C
CHARCOT-MARIE-TOOTH DISEASE WITH DEAFNESS, X-LINKED (CMTX1) (*GJB1*) **negative**
CHOROIDEREMIA (*CHM*) **negative**
CHRONIC GRANULOMATOUS DISEASE, X-LINKED (*CYBB*) **negative**
COWCHOCK SYNDROME (*AIFM1*) **negative**
CREATINE TRANSPORTER DEFECT (Cerebral Creatine Deficiency Syndrome 1, X-Linked) (*SLC6A8*) **negative**

D
DENT DISEASE, TYPE 1 (*CLCN5*) **negative**
DENT DISEASE, TYPE 2 / LOWE SYNDROME (*OCRL*) **negative**
DUCHENNE / BECKER MUSCULAR DYSTROPHY (*DMD*) **negative**
DYSKERATOSIS CONGENITA, DKC1-RELATED (*DKC1*) **negative**

E
EMERY-DREIFUSS MUSCULAR DYSTROPHY 1, X-LINKED (*EMD*) **negative**

F
FABRY DISEASE (*GLA*) **negative**
FACTOR IX DEFICIENCY (*F9*) **negative**
FANCONI ANEMIA, GROUP B (*FANCB*) **negative**
FRAGILE X SYNDROME (*FMR1*) **negative 29 and 30 CGG repeats were detected in the FMR1 genes.**

G
GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY (*G6PD*) **negative**

H
HETEROTAXY SYNDROME, ZIC3-RELATED (*ZIC3*) **negative**
HYPER IGM SYNDROME, X-LINKED (*CD40LG*) **negative**
HYPOHIDROTIC ECTODERMAL DYSPLASIA, X-LIINKED (*EDA*) **negative**

I
IMMUNE DYSREGULATION, POLYENDOCRINOPATHY, ENTEROPATHY, X-LINKED (IPEX) SYNDROME (*FOXP3*) **negative**
INFANTILE SPINAL MUSCULAR ATROPHY, X-LINKED (*UBA1*) **negative**
ISOLATED LISSENCEPHALY SEQUENCE/SUBCORTICAL BAND HETEROTOPIA (*DCX*) **negative**

J
JUVENILE RETINOSCHISIS, X-LINKED (*RS1*) **negative**

L
L1 SYNDROME (*L1CAM*) **negative**
LESCH-NYHAN SYNDROME (*HPRT1*) **negative**

M
MENKES SYNDROME (*ATP7A*) **negative**
MUCOPOLYSACCHARIDOSIS, TYPE II (HUNTER SYNDROME) (*IDS*) **negative**
MYOTUBULAR MYOPATHY, X-LINKED (*MTM1*) **negative**

O
ORNITHINE TRANSCARBAMYLASE DEFICIENCY (*OTC*) **negative**

P
PYRUVATE DEHYDROGENASE DEFICIENCY, X-LINKED (*PDHA1*) **negative**

S
SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED (*IL2RG*) **negative**

W
WISKOTT-ALDRICH SYNDROME (*WAS*) **negative**

X
X-LINKED LISSENCEPHALY WITH ABNORMAL GENITALIA (*ARX*) **negative**

Patient Information		Test Information	
Patient Name:	Elizabeth Bennett	Ordering Physician:	N/A
		Clinic Information:	Natera, Inc.
Date Of Birth:	01/12/1996		
Case File ID:	3113050		
		Report Date:	01/19/2021

Testing Methodology, Limitations, and Comments:

Next-generation sequencing (NGS)

1. For the paired-end pre-capture library procedure, genomic DNA is fragmented by sonication and ligated to the Illumina multiplexing PE adapters with sequencing barcodes (indexes). The adapter-ligated DNA is then PCR amplified using primers. For the target enrichment capture procedure, the pre-capture libraries are pooled as a 47-plex and enriched by hybridizing to biotin-labeled probe (smallUTCv3) in-solution at 56C or 47C for 16-48 hours. The post-capture library DNA is subjected to massively parallel sequencing on the Illumina HiSeq 2500 platform for 100 bp paired-end reads. The following quality control metrics of the sequencing data are generally achieved: >98% target bases covered at >20X, >95% target bases covered at >40X, mean coverage of target bases at >120X. SNP concordance to genotype array: >95%. This test may not provide detection of a portion of the gene due to local sequence characteristics or the presence of closely related pseudogenes. Terminal deletions and duplications may not be fully delineated. Partial exonic copy number changes and sequences present in repetitive sequences may not be identified by this methodology.
2. As a quality control measure, analysis includes a genotyping assay performed by the Fluidigm SNPtype platform using the SNPTrace Panel. The SNPTrace Panel consists of 90 autosomal loci and 6 allosome loci (3 SNPs each on chrX and chrY). Samples and assays are transferred to a 96 Fluidigm Dynamic array, loaded to reaction chambers by an integrated fluidic circuit (IFC) controller, thermal cycled, and endpoint-imaged on the BioMark HD System (Fluidigm). The SNP data are first analyzed by SNP Genotyping Analysis Software (Fluidigm) and then by comparison with the genotype calls made from the Dragen BioIT Platform for NGS data to ensure correct sample identification. Once an assessment of identity match is established, contamination analysis is performed by using homozygous sites and computational inspection of BAM data.
3. Data analysis and interpretation are performed by the Baylor Genetics analytics pipeline. The output data from the Illumina HiSeq are converted from BCL files to FastQ files according to each sample's specific adapter sequence using Illumina's recommended procedure. FastQ data are aligned to the human reference genome using the Dragen BioIT Platform (Illumina). The output of the alignment is a BAM file; QC metrics of the map-align process are recorded for quality review. QC statistics include coverage for target genes and known pathogenic variant sites, mate-pair alignment information as well as number of total and duplicate reads. Variant calling on the BAM file is performed using the Dragen haplotype-based variant calling system. The variant calling step generates a "raw" VCF file containing a list of detected variants, which are then annotated using a locally installed annotation system. The annotation platform leverages the GenomOncology Knowledge Management System API and provides annotations using open source data sets such as ExAC, EVS, and ClinVar and professional resources such as HGMD Pro. The API also provides HGVS nomenclature built using the Biocommons open-source suite of tools: HGVS python library, the UTA transcript repository, and SeqRepo sequence database. This annotation system reports zygosity as well as inference of mutation types including nonsense, missense, synonymous, splicing and frameshift, among others. Synonymous variants, intronic variants not affecting splicing sites, and common benign variants are excluded from interpretation unless previously reported as pathogenic variants. It should be noted that the data interpretation is based on our current understanding of the gene and variants at the time of reporting. The sequence alignment, variant calling and annotation algorithms may be updated periodically with validated improvements and increments to the knowledgebase.
4. Copy number variants (CNV) are analyzed by the Baylor Genetics analytics pipeline. CNV analysis is limited to deletions involving more than one exon for most genes in the panel, except specific known recurrent deletion events, and exonic deletion and multi-exonic duplication events of CFTR, DMD, and HBB. The method does not detect gene inversions, most single-exonic deletions, and duplications. Additionally, the method does not define the exact deletion/duplication boundaries 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 methods for confirmation and variant sizing; additional information.
5. SMN1 exon 7 deletion and g.27134T>G SNP analysis is performed by the Baylor Genetics analytics pipeline. The SMN1 analysis does not identify a carrier who has an exon 7 deletion on one chromosome and two copies of the SMN1 gene on the other chromosome.
6. If CYP21A2 is tested, CYP21A2 deletion and sequencing variant analysis is performed by the Baylor Genetics analytics pipeline. Specific enhancement on variant detection is applied to c.92C>T (p.P31L), c.293-13C>G, c.332_339delGAGACTAC (p.G111Vfs*21), c.518T>A (p.I173N), c.(710T>A;713T>A)(I237N;V238E), c.844G>T (p.V282L), c.923dupT (p.L308Ffs*6), c.955C>T (p.Q319*), c.1069C>T (p.R357W), c.1360C>T (p.P454S). CYP21A2 duplication will be reflexed if c.955C>T (p.Q319*) is detected. Sequencing variants outside these regions might be detected as well.

Reflex Testing

If the CFTR R117H variant is detected, reflex testing of the polythymidine variations (5T, 7T and 9T) at the intron 9 (legacy intron number 8) branch/acceptor site of the CFTR gene will be performed. The polythymidine variations (5T, 7T and 9T) are analyzed by Sanger sequencing.

Sanger Sequencing

A PCR-based assay is used to amplify the region(s) of interest in the gene. Direct sequence analysis of PCR products is performed in both the forward and reverse directions using automated fluorescence dideoxy sequencing methods.

Patient Information

Patient Name: Elizabeth Bennett

Date Of Birth: 01/12/1996

Case File ID: 3113050

Test Information

Ordering Physician: N/A

Clinic Information: Natera, Inc.

Report Date: 01/19/2021

Fragile X

The CGG repeat region at the FMR1 5'-untranslated region is amplified by fluorescent PCR followed by capillary electrophoresis. Allele sizes up to 200 repeats are analyzed by GeneMapper software. Reflex Southern blot analysis will be performed for samples with full mutation allele in order to assess size and methylation status for larger repeats. This analysis does not detect deletions or point mutations, which comprises less than one percent of the FMR1 pathogenic variants. Reflex testing for number of AGG interruptions is performed for all CGG repeat sizes between 55 and 90. A separate report will be issued with the AGG results. AGG interruption testing is performed by Asuragen, Inc., 2150 Woodward St. Suite 100 Austin, TX 78744 (CLIA ID: 45D1069375).

Fragile X Repeat Categories

Categories	**CGG Repeat Sizes**
Normal	<45
Intermediate	45 - 54
Premutation	55 - 200
Full	>200

Spinal Muscular Atrophy

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 of 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