NTD | Eurofins Clinical Diagnostics

MM470: Pan-Ethnic Carrier Screen - Gene Sequencing Panel

Report Prepared For:

Patient:

Name: [redacted]
Date of Birth: [redacted]
IDs (Internal / External): [redacted]
Reported Gender: [redacted]
Reported Ethnicity: [redacted]

Physician/Institution Referral:

Physician Information

Institution Information
Institution Address

Sample:

Sample Collection Date: [redacted]
Sample Received Date: [redacted]
Final Report Date: [redacted]
Sample Type: [redacted]
IDs (Internal / External): [redacted]

Report Summary:

Autosomal Carrier Screening Results: **Positive Carrier**

This individual tested positive as a carrier of the following disease(s).

Carnitine Palmitoyltransferase II Deficiency

CPT2:NM_000098.2:c.370C>T (p.Arg124*)

X-Linked Carrier Screening Results: **Negative**

This individual tested negative for pathogenic variants in the X-linked genes analyzed. Residual carrier risks for this individual based on reported ethnicity are given in a following table. These results must be interpreted in the context of this individual's clinical profile and family history. Genetic counseling is recommended.

Fragile X Repeat Analysis: **Premutation**

An FMR1 gene expansion in the premutation range (55-200 repeats) was detected in this individual. While carriers of premutations are unaffected by fragile X syndrome, adult men carrying an FMR1 premutation have an elevated risk of developing fragile X associated tremor/ataxia syndrome (FXTAS), which is characterized by late-onset (after age 50), progressive loss of muscle coordination, tremor, and short-term memory loss. Female relatives of this individual, if they carry the FMR1 premutation, are at risk of transmitting an expanded fragile X full mutation to their children and having children affected with fragile X syndrome. Adult female carriers of FMR1 premutation are at elevated risk of premature ovarian insufficiency.

FMR1 gene CGG Repeats	100

Spinal Muscular Atrophy (SMA) Analysis: **Negative**

Two copies of the SMN1 gene were identified. While having 2 copies reduces the chance of being a carrier, this risk is not zero and will vary by ethnicity.

Alpha Thalassemia Analysis: **Positive**

Analysis of the alpha-globin genes HBA1 and HBA2 detected one copy of a SEA deletion. This deletion removes both alpha 1 and alpha 2 genes from a single chromosome. Loss of alpha 1 and alpha 2 globin genes is consistent with a carrier of the SEA alpha-globin gene deletion (aa/--).

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Other Findings of Note: [redacted] **Other** [redacted]

Metachromatic Leukodystrophy

ARSA:NM_000487.5:c.*96A>G

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Positive Carrier Result Details:

Carnitine Palmitoyltransferase II Deficiency **CPT2**

Carnitine palmitoyltransferase II deficiency prevents the body from using fat for energy, so the fuel they need to function properly is lessened, especially in times of fasting, exercise, or infection. People with this condition have low blood sugar, an enlarged, malfunctioning liver, and often seizures. This condition also weakens the heart muscle and cause breathing difficulties. There is a range of severity, with the more severe forms having a reduced lifespan.

This condition is inherited in an autosomal recessive manner which means both parents have to be carriers to have a 1 in 4 (25%) risk to have an affected child. Being a carrier does not mean you are affected with this condition, but it can increase the risk for you to have affected children. These risks will vary depending on the carrier status of your partner, and it is recommended that you discuss these results and the available follow-up testing options with your healthcare provider. You may also wish to share your results with other family members so they can consider testing as their risks for being a carrier is also increased.

Variants Detected:

CPT2:NM_000098.2:c.370C>T (p.Arg124*)

Prior Reproductive Risk: 1 in 1,000,000 *(Specific to partners of reported ethnicity prior to testing)*

Current Reproductive Risk: <see below> *(Please see table below)*

Current Reproductive Risk:

Partner Ethnicity:	Partner Is Untested: (1)	Partner is Negative: (1, 2)	Partner is a Carrier:
African American	1 in 2,000	1 in 40,000	1 in 4
Asian	1 in 2,000	1 in 40,000	1 in 4
European / Caucasian	1 in 2,000	1 in 40,000	1 in 4
Finnish	1 in 2,000	1 in 40,000	1 in 4
Hispanic	1 in 2,000	1 in 40,000	1 in 4
Ashkenazi Jewish	1 in 2,000	1 in 40,000	1 in 4
Other / Mixed	1 in 2,000	1 in 40,000	1 in 4

1 - Based upon the partner's general population carrier rate for the given ethnicity (available upon request).

2 - Based upon the partner testing negative for the mutations on this carrier screen and the detection rate for the ethnicity given (available upon request).

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Affected / Positive Carrier Result Details:

Metachromatic Leukodystrophy **ARSA**

The protective covering of the nerve cells is damaged in people with metachromatic leukodystrophy. People with this condition experience damage to the muscles over time where they become severely weakened and eventually rigid. Loss of sensation in the limbs is also common, and many experience vision and hearing loss as well as seizures. Although the majority of people with this condition show signs in infancy, childhood and adult forms have also been reported. Behavioral changes and psychiatric illnesses such as hallucinations can occur in the later onset forms. All forms progress to a stage of paralysis and unresponsiveness with shortened lifespan.

*This individual carries two copies of the c.*96A>G change. The specific change is known to be a pseudodeficiency allele. A pseudodeficiency allele is a DNA change that is not powerful enough to cause carrier status and increase the risk for having a child with MLD, but it has been shown to give a misleading result on biochemical studies. Biochemical testing of this individual or her/his child who inherits this change may give abnormal results even though the individual does not have MLD. This individual was not found to be a carrier of a mutation on this panel that could cause MLD in her/his children.*

Variants Detected:

ARSA:NM_000487.5:c.*96A>G

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Residual Carrier Risks:

The following prior risks are based upon incidence/carrier rates published in the scientific literature, the patient's reported ethnicity, and that the patient does not have a family history of the disease. The residual risks are based upon the disease-specific detection rates and reflect remaining risk to be a carrier after a negative screening result. Residual risks in males for X-linked conditions will not be displayed as the reproductive risk falls primarily with the female partner. For more information or for calculation of reproductive risks in various partner-testing scenarios, please refer to our website at http://eglgenetics.com/pecs/.

The incidence/carrier rates are based on our current understanding of the conditions and genes on this panel. These rates may change over time as more information about the conditions and genes and their incidence in the general population becomes available.

	Carrier Risk:	
Condition (Gene):	*Prior:*	**Residual:**
Hyperinsulinism (ABCC8)	*1 in 110*	<1 in 120
Medium Chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	*1 in 65*	<1 in 640
Short Chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	*1 in 100*	<1 in 110
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	*1 in 87*	<1 in 110
Beta-Ketothiolase Deficiency (ACAT1)	*1 in 500*	<1 in 560
Aspartylglycosaminuria (AGA)	*1 in 500*	<1 in 560
Glycogen Storage Disease Type III (Cori/Forbes) (AGL)	*1 in 160*	<1 in 220
Hyperoxaluria, Primary Type 1 (AGXT)	*1 in 170*	<1 in 350
Polyglandular Autoimmune Syndrome Type 1 (AIRE)	*1 in 500*	<1 in 1,500
Sjogren-Larsson Syndrome (ALDH3A2)	*1 in 500*	<1 in 660
Hereditary Fructose Intolerance (ALDOB)	*1 in 71*	<1 in 470
Hypophosphatasia (ALPL)	*1 in 160*	<1 in 230
Mucopolysaccharidosis Type VI (Maroteaux-Lamy) (ARSB)	*1 in 250*	<1 in 340
Argininosuccinate Lyase Deficiency (ASL)	*1 in 130*	<1 in 180
Canavan Disease (ASPA)	*1 in 500*	<1 in 1,000
Citrullinemia Type 1 (ASS1)	*1 in 120*	<1 in 200
Ataxia-Telangiectasia (ATM)	*1 in 100*	<1 in 110
Wilson Disease (ATP7B)	*1 in 87*	<1 in 130
Bardet-Biedl Syndrome, Type 1 (BBS1)	*1 in 330*	<1 in 1,600
Bardet-Biedl Syndrome, Type 10 (BBS10)	*1 in 350*	<1 in 650
Maple Syrup Urine Disease Type 1A (BCKDHA)	*1 in 320*	<1 in 360
Maple Syrup Urine Disease Type 1B (BCKDHB)	*1 in 360*	<1 in 400
GRACILE Syndrome (BCS1L)	*1 in 500*	<1 in 560
Bloom Syndrome (BLM)	*1 in 500*	<1 in 560
Biotinidase Deficiency (BTD)	*1 in 120*	<1 in 300
Limb-Girdle Muscular Dystrophy Type 2A (CAPN3)	*1 in 160*	<1 in 180
Homocystinuria, CBS-deficient (CBS)	*1 in 250*	<1 in 370
Cystic Fibrosis (CFTR)	*1 in 25*	<1 in 340
Neuronal Ceroid Lipofuscinosis Type 3 (CLN3)	*1 in 160*	<1 in 580
Neuronal Ceroid Lipofuscinosis Type 5 (CLN5)	*1 in 160*	<1 in 830
Neuronal Ceroid Lipofuscinosis Type 8 (a.k.a Northern Epilepsy) (CLN8)	*1 in 500*	<1 in 560
Usher Syndrome Type 3 (CLRN1)	*1 in 500*	<1 in 560
Achromatopsia (CNGB3)	*1 in 410*	<1 in 680
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	*1 in 500*	<1 in 560
Cystinosis (CTNS)	*1 in 160*	<1 in 320
Papillon-Lefevre Syndrome (also Haim-Munk Syndrome) (CTSC)	*1 in 500*	<1 in 560
Pycnodysostosis (CTSK)	*1 in 150*	<1 in 170
Glaucoma, Primary Congenital (CYP1B1)	*1 in 87*	<1 in 96
Congenital Adrenal Hyperplasia (CAH) (CYP21A2)	*1 in 61*	<1 in 68
Maple Syrup Urine Disease Type 2 (DBT)	*1 in 480*	<1 in 530
Smith-Lemli-Opitz Syndrome (DHCR7)	*1 in 71*	<1 in 230
Dihydrolipoamide Dehydrogenase Deficiency (a.k.a Maple Syrup Urine Disease Type 3) (DLD)	*1 in 500*	<1 in 560

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Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	*1 in 500*	<1 in 1,000
Hypohidrotic Ectodermal Dysplasia (EDAR)	*1 in 65*	<1 in 72
Familial Dysautonomia (ELP1)	*1 in 500*	<1 in 560
Factor XI Deficiency (F11)	*1 in 500*	<1 in 560
Tyrosinemia Type I (FAH)	*1 in 140*	<1 in 340
Fanconi Anemia Type C (FANCC)	*1 in 300*	<1 in 330
Fumarase Deficiency (FH)	*1 in 500*	<1 in 710
Walker- Warburg Syndrome, Type 4 (FKTN)	*1 in 500*	<1 in 560
Glycogen Storage Disease Type Ia (von Gierke) (G6PC)	*1 in 180*	<1 in 610
Pompe Disease (a.k.a Glycogen Storage Disease Type 2 or Acid Maltase Deficiency) (GAA)	*1 in 160*	<1 in 630
Krabbe Disease (GALC)	*1 in 160*	<1 in 630
Mucopolysaccharidosis Type IVA (Morquio A) (GALNS)	*1 in 230*	<1 in 290
Galactosemia (GALT)	*1 in 110*	<1 in 360
Gaucher Disease (GBA)	*1 in 120*	<1 in 400
Glutaric Acidemia Type 1 (GCDH)	*1 in 110*	<1 in 200
Growth Hormone Deficiency, Isolated (GHRHR)	*1 in 50*	<1 in 55
Hearing Loss, Non-syndromic (a.k.a Connexin 26) (GJB2)	*1 in 42*	<1 in 230
Hearing Loss, Non-syndromic (a.k.a Connexin 30) (GJB6)	*1 in 150*	<1 in 15,000
Mucopolysaccharidosis Type IVB (Morquio B) (GLB1)	*1 in 500*	<1 in 560
Inclusion Body Myopathy 2 (GNE)	*1 in 500*	<1 in 560
Mucolipidosis Type II/IIIA (GNPTAB)	*1 in 160*	<1 in 290
Bernard-Soulier Syndrome, Type B (GP1BB)	*1 in 500*	<1 in 560
Bernard-Soulier Syndrome,Type C (GP9)	*1 in 500*	<1 in 560
Hyperoxaluria, Primary Type 2 (GRHPR)	*1 in 500*	<1 in 1,100
Mucopolysaccharidosis Type VII (Sly) (GUSB)	*1 in 250*	<1 in 280
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (HADHA)	*1 in 500*	<1 in 1,700
Beta-Hemoglobinopathies (including Sickle Cell and Beta-Thalassemia) (HBB)	*1 in 32*	<1 in 180
Tay Sachs Disease (a.k.a. Hexosaminidase A Deficiency) (HEXA)	*1 in 300*	<1 in 750
Sandhoff Disease (HEXB)	*1 in 500*	<1 in 680
Hemochromatosis (HFE)	*1 in 9*	<1 in 90
Heme Oxygenase 1 Deficiency (HMOX1)	*1 in 500*	<1 in 560
D-Bifunctional Protein Deficiency (HSD17B4)	*1 in 160*	<1 in 180
Mucopolysaccharidosis Type I (Hurler) (IDUA)	*1 in 190*	<1 in 680
Isovaleric Acidemia (IVD)	*1 in 250*	<1 in 280
Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related (LAMA3)	*1 in 500*	<1 in 910
Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related (LAMB3)	*1 in 500*	<1 in 910
Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related (LAMC2)	*1 in 500*	<1 in 910
Woolly Hair/Hypotrichosis Syndrome (LIPH)	*1 in 500*	<1 in 560
Alpha-Mannosidosis (MAN2B1)	*1 in 270*	<1 in 370
Mucolipidosis Type IV (MCOLN1)	*1 in 500*	<1 in 560
Familial Mediterranean Fever (MEFV)	*1 in 500*	<1 in 560
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	*1 in 500*	<1 in 560
Methlymalonic Acidemia, cblA Type (MMAA)	*1 in 280*	<1 in 500
Methlymalonic Acidemia, cblB-Type (MMAB)	*1 in 410*	<1 in 610
Methylmalonic Acidemia and Homocystinuria, cblC Type (MMACHC)	*1 in 500*	<1 in 830
Congenital Disorder of Glycosylation Type Ib (MPI)	*1 in 500*	<1 in 560
Methlymalonic Acidemia, mut-Type (MUT)	*1 in 180*	<1 in 200
Mucopolysaccharidosis Type IIIB (Sanfilippo B) (NAGLU)	*1 in 210*	<1 in 240
Nijmegen Breakage Syndrome (NBN)	*1 in 160*	<1 in 520
Nemaline Myopathy (NEB)	*1 in 110*	<1 in 120
Hydatidiform Mole, Recurrent (NLRP7)	*1 in 500*	<1 in 560
Niemann-Pick Disease Type C1 (NPC1)	*1 in 160*	<1 in 190

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Niemann-Pick Disease Type C2 (NPC2)	*1 in 500*	<1 in 760
Finnish Nephrosis (a.k.a Nephrotic Syndrome Type 1) (NPHS1)	*1 in 500*	<1 in 560
Nephrotic Syndrome Type 2 (NPHS2)	*1 in 500*	<1 in 750
Costeff Optic Atrophy Syndrome (OPA3)	*1 in 500*	<1 in 560
Phenylalanine Hydroxylase Deficiency (PKU) (PAH)	*1 in 50*	<1 in 110
Pantothenate Kinase-associated Neurodegeneration (PANK2)	*1 in 500*	<1 in 560
Usher Syndrome Type 1F (PCDH15)	*1 in 270*	<1 in 300
Zellweger Spectrum Disorder Type 1 (a.k.a Infantile Refsum Disease) (PEX1)	*1 in 110*	<1 in 560
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	*1 in 160*	<1 in 480
Polycystic Kidney Disease, Autosomal Recessive (PKHD1)	*1 in 70*	<1 in 78
Congenital Disorder of Glycosylation Type Ia (PMM2)	*1 in 71*	<1 in 250
Muscle-Eye-Brain Disease (POMGNT1)	*1 in 500*	<1 in 560
Neuronal Ceroid Lipofuscinosis Type 1 (PPT1)	*1 in 160*	<1 in 340
Combined Pituitary Hormone Deficiency (PROP1)	*1 in 45*	<1 in 130
Glycogen Storage Disease Type V (McArdle) (PYGM)	*1 in 160*	<1 in 1,600
Cartilage-Hair Hypoplasia (RMRP)	*1 in 500*	<1 in 960
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS) (SACS)	*1 in 500*	<1 in 560
Alpha-1 Antitrypsin Deficiency (SERPINA1)	*1 in 35*	<1 in 690
Limb-Girdle Muscular Dystrophy Type 2D (SGCA)	*1 in 280*	<1 in 410
Limb-Girdle Muscular Dystrophy Type 2E (SGCB)	*1 in 500*	<1 in 560
Limb-Girdle Muscular Dystrophy Type 2C (SGCG)	*1 in 410*	<1 in 460
Mucopolysaccharidosis Type IIIA (Sanfilippo A) (SGSH)	*1 in 170*	<1 in 240
Andermann Syndrome (SLC12A6)	*1 in 500*	<1 in 560
Salla Disease (a.k.a Sialic Acid Storage Disease) (SLC17A5)	*1 in 500*	<1 in 560
Megaloblastic Anemia Syndrome (SLC19A2)	*1 in 500*	<1 in 560
Carnitine Deficiency, Primary (SLC22A5)	*1 in 110*	<1 in 120
Skeletal Dysplasias, SLC26A2-related (SLC26A2)	*1 in 160*	<1 in 520
Pendred Syndrome (SLC26A4)	*1 in 50*	<1 in 99
Glycogen Storage Disease Type Ib (von Gierke) (SLC37A4)	*1 in 350*	<1 in 650
Niemann-Pick Disease Type A & B (a.k.a. Acid Sphingomyelinase Deficiency) (SMPD1)	*1 in 160*	<1 in 230
Segawa Syndrome (TH)	*1 in 500*	<1 in 560
Joubert Syndrome 2 (TMEM216)	*1 in 500*	<1 in 560
Neuronal Ceroid Lipofuscinosis Type 2 (TPP1)	*1 in 160*	<1 in 390
Tricho-Hepato-Enteric Syndrome (TTC37)	*1 in 500*	<1 in 560
Ataxia With Vitamin E Deficiency (TTPA)	*1 in 500*	<1 in 560
Oculocutaneous Albinism Type 1 (TYR)	*1 in 100*	<1 in 110
Cohen Syndrome (VPS13B)	*1 in 500*	<1 in 560
Progressive Pseudorheumatoid Dysplasia (WISP3)	*1 in 500*	<1 in 560
Werner Syndrome (WRN)	*1 in 220*	<1 in 250

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Test Methodology:

Full gene sequence analysis: In solution hybridization of all coding exons within the genes tested including large indels was performed on this individual's genomic DNA. Direct sequencing of the amplified captured regions was performed using next generation short base pair read sequencing. Please note, sequence analysis may not be completed for those genes which have pseudogenes. Nucleotide numbering is based on GenBank accession numbers given in the report and on the EGL Genetics website; nucleotide 1 corresponds to the A of the start codon ATG.

Targeted deletion/duplication analysis: DNA isolated from peripheral blood is hybridized to a comparative genomic hybridization (CGH) array to detect deletions and duplications. The targeted CGH array is custom designed for analyzing the CFTR, DMD, and MECP2 genes. Rarely, probe coverage may be limited or absent in some exons due to the features of the targeted sequence. Please note, this assay was designed to detect whole gene deletions for the globin genes. Genomic coordinate numbering is based on GRCh37/hg19.

Alpha thalassemia analysis: DNA isolated from this individual was evaluated for copy number changes in the HBA1 and HBA2 genes using multiplex ligation polymerase chain reaction amplification (MLPA) according to the SALSA protocol available from MRC Holland. Copy number changes analyzed by this assay include the -α3.7,- α4.2,-(α)20.5,--SEA,--MED,--FIL, and –THAI alpha-globin gene deletions. The gene dosage ratio was calculated relative to the average of 11 reference loci. Two copies of the HBA1 and HBA2 genes most likely indicate normal (not affected) status and deletions of this region most likely indicate affected or carrier status. Note: Sequence changes and other deletions in these genes not targeted by this assay are not detected by this analysis.

Fragile X syndrome CGG repeat analysis: The DNA surrounding the CGG repeat in the FMR1 gene was amplified by PCR and the size of the repeat was determined by capillary electrophoresis. Normal: <45 CGG repeats, Intermediate: 45-54 CGG repeats, Premutation: 55-~200 repeats, Full mutation: >200 CGG repeats (ACMG Standards and Guidelines for fragile X testing. Monaghan et al. Genet Med 2013:15(7):575–586). Other types of mutations in the FMR1 gene will not be detected by this assay. Methylation status is not indicated by this assay. Low levels of mosaicism may not be detected.

Spinal muscular atrophy deletion analysis: Isolated DNA from this individual was evaluated for SMN1 gene deletions using multiplex ligation polymerase chain reaction amplification (MLPA) of exons 7 and 8 according to the SALSA protocol available from MRC Holland. The gene dosage ratio was calculated relative to the average of 16 reference loci. Two copies of SMN1 most likely indicates normal (not affected) status and one copy of SMN1 most likely indicates carrier status. Note: Other pathogenic variants in SMN1, such as small pathogenic variants and fusion SMN genes, are possible in this gene but are not detected by this assay. Additionally a duplication of SMN1 on one chromosome may interfere with the detection of a deletion of SMN1 on the opposite chromosome.

Sanger sequencing: In order to avoid pseudogene sequence regions, targeted GBA mutations were analyzed by Sanger sequence analysis. The targeted mutations include: c.84dupG, c.115+1G>A, c.1226A>G (p.N409S), c.1263_1317del55, c.1297G>T (p.V433L), c.1342G>C (p.D448H), c.1343A>T (p.D448V), c.1448T>C (p.L483P), c.1504C>T (p.R502C), c.1505G>A (p.R502H) and c.1604G>A (p.R535H). PCR was used to amplify the targeted mutation regions. The PCR products were sequenced bidirectionally. Nucleotide numbering is based on GenBank accession numbers given in the report and on the EGL Genetics website; nucleotide 1 corresponds to the A of the start codon ATG.

Version AT MLPA, DDPECSV02, MFRAX, NG470V12, PECS_GBA, SMACAR

Disclaimer:

Because of the nature of X-linked inheritance, this test, if positive, may be diagnostic for male patients in rare cases. These results must be interpreted in the context of this individual's clinical profile and family history. This analysis does not include testing for all mutations found in these genes. Negative results therefore reduce the risk of being a carrier but do not eliminate it. Further testing may be indicated based on family history or ethnicity, such as CBC and hemoglobin electrophoresis in those of African, Southeast Asian, and Mediterranean descent, or Tay-Sachs disease carrier analysis by enzyme. Possible diagnostic errors include sample mix-ups, genetic variants that interfere with analysis, and other sources.

Testing was performed and results originated from EGL Genetic Diagnostics LLC (CLIA#:11D0683478; CAP#: 7181693, Lora J. H. Bean, PhD, FACMG, Director), 2460 Mountain Industrial Boulevard, Tucker, GA 30084. Pursuant to the requirements of CLIA 1988, this test was developed and its performance validated by EGL Genetic Diagnostics LLC. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes.

This case has been reviewed and electronically signed by a Laboratory Director at EGL Genetics.

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