NTD | Eurofins Clinical Diagnostics

MM480: Pan-Ethnic Carrier Screen - Targeted Mutation Panel

Report Prepared For:

Patient:

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

Sample:

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

Physician/Institution Referral:

[redacted]
Institution Name
Institution Address

[redacted] [redacted] [redacted]
Physician's Name
Physicain's Address

Report Summary:

Autosomal Carrier Screening Results: **Positive Carrier**

This individual tested positive as a carrier of the following disease(s). None of the other autosomal recessive mutations on the panel were detected. Mutations other than those on the panel will not be detected.

Ataxia-Telangiectasia

ATM:NM_000051.3:c.7638_7646delTAGAATTTC (p.Arg2547_Ser2549del)

Smith-Lemli-Opitz Syndrome

DHCR7:NM_001360.2:c.1055G>A (p.Arg352Gln)

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

This individual tested negative for all X-linked mutations 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. This panel does not analyze all possible mutations in the genes included in this assay. This individual may still carry a pathogenic variant not included in this panel. Genetic counseling is recommended.

Fragile X Repeat Analysis: **Normal**

The common FMR1 gene repeat expansion associated with fragile X syndrome was not detected. Other types of mutations in the FMR1 gene will not be detected by this assay. This result significantly reduces but does not eliminate the chance of this individual being a carrier of fragile X syndrome.

FMR1 gene CGG Repeats 29 / 30

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

The common SMN1 gene deletion associated with spinal muscular atrophy (SMA) was detected. This result indicates that this individual is a carrier of SMA. 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.

Alpha Thalassemia Analysis: **Positive**

One copy of a 3.7 deletion (-a3.7) was detected in this individual. This is a single alpha-globin gene deletion. Loss of a single alphaglobin gene is consistent with alpha-thalassemia silent carrier (aa/a-).

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Positive Carrier Result Details:

Ataxia-Telangiectasia **ATM**

Ataxia-telangiectasia is a condition that results from increased DNA damage and abnormal cell division. People with this condition have problems with coordination, often have muscle twitches, high rates of infection, and enlarged blood vessels. They are extremely sensitive to X-ray exposure.

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:

ATM:NM_000051.3:c.7638_7646delTAGAATTTC (p.Arg2547_Ser2549del)

Prior Reproductive Risk: 1 in 40,000 *(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 400	1 in 440	1 in 4
Asian	1 in 400	1 in 440	1 in 4
European / Caucasian	1 in 400	1 in 440	1 in 4
Finnish	1 in 400	1 in 440	1 in 4
Hispanic	1 in 400	1 in 440	1 in 4
Ashkenazi Jewish	1 in 400	1 in 440	1 in 4
Other / Mixed	1 in 400	1 in 440	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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Positive Carrier Result Details:

Smith-Lemli-Opitz Syndrome **DHCR7**

Smith-Lemli-Opitz syndrome (SLOS) causes the body to be unable to process cholesterol, which is important to many areas of the body. Children with SLOS grow slowly, have weak muscles, feeding difficulties, and over 90% will have some degree of intellectual disability. Cleft palate and abnormalities of the hands and/or feet are also common. The build-up of cholesterol also affects the kidneys and heart. Although some of these effects can be severe and lead to early death, proper medical attention and diet can often lengthen the lifespan of people with this condition.

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:

DHCR7:NM_001360.2:c.1055G>A (p.Arg352Gln)

Prior Reproductive Risk: 1 in 20,000 *(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 280	1 in 900	1 in 4
Asian	1 in 280	1 in 900	1 in 4
European / Caucasian	1 in 280	1 in 900	1 in 4
Finnish	1 in 280	1 in 900	1 in 4
Hispanic	1 in 280	1 in 900	1 in 4
Ashkenazi Jewish	1 in 280	1 in 900	1 in 4
Other / Mixed	1 in 280	1 in 900	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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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 180
Hyperoxaluria, Primary Type 1 (AGXT)	*1 in 500*	1 in 1,000
Polyglandular Autoimmune Syndrome Type 1 (AIRE)	*1 in 500*	1 in 1,500
Sjogren-Larsson Syndrome (ALDH3A2)	*1 in 500*	1 in 560
Hereditary Fructose Intolerance (ALDOB)	*1 in 71*	1 in 78
Hypophosphatasia (ALPL)	*1 in 160*	1 in 230
Metachromatic Leukodystrophy (ARSA)	*1 in 100*	1 in 110
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
Wilson Disease (ATP7B)	*1 in 87*	1 in 96
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 330
Cystic Fibrosis (CFTR)	*1 in 61*	1 in 320
Choroideremia (CHM)	*1 in 25,000*	1 in 28,000
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
Carnitine Palmitoyltransferase II Deficiency (CPT2)	*1 in 500*	1 in 10,000
Cystinosis (CTNS)	*1 in 160*	1 in 180
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

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Dihydrolipoamide Dehydrogenase Deficiency (a.k.a Maple Syrup Urine Disease Type 3) (DLD)	*1 in 500*	1 in 560
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
Hemophilia B (F9)	*1 in 20,000*	1 in 24,000
Tyrosinemia Type I (FAH)	*1 in 140*	1 in 150
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
Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD)	*1 in 10*	1 in 12
Pompe Disease (a.k.a Glycogen Storage Disease Type 2 or Acid Maltase Deficiency) (GAA)	*1 in 59*	1 in 230
Krabbe Disease (GALC)	*1 in 160*	1 in 180
Mucopolysaccharidosis Type IVA (Morquio A) (GALNS)	*1 in 230*	1 in 290
Galactosemia (GALT)	*1 in 110*	1 in 120
Gaucher Disease (GBA)	*1 in 120*	1 in 400
Glutaric Acidemia Type 1 (GCDH)	*1 in 110*	1 in 120
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 87
Hearing Loss, Non-syndromic (a.k.a Connexin 30) (GJB6)	*1 in 150*	1 in 15,000
Fabry Disease (GLA)	*1 in 50,000*	1 in 56,000
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 560
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 5*	1 in 19
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 13*	1 in 130
Heme Oxygenase 1 Deficiency (HMOX1)	*1 in 500*	1 in 560
D-Bifunctional Protein Deficiency (HSD17B4)	*1 in 160*	1 in 180
Mucopolysaccharidosis Type II (Hunter) (IDS)	*1 in 100,000*	1 in 110,000
Mucopolysaccharidosis Type I (Hurler) (IDUA)	*1 in 190*	1 in 630
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 310
Methlymalonic Acidemia, cblB-Type (MMAB)	*1 in 410*	1 in 450
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

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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
Niemann-Pick Disease Type C2 (NPC2)	*1 in 500*	1 in 560
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 560
Costeff Optic Atrophy Syndrome (OPA3)	*1 in 500*	1 in 560
Ornithine Transcarbamlyase Deficiency (OTC)	*1 in 35,000*	1 in 39,000
Phenylalanine Hydroxylase Deficiency (PKU) (PAH)	*1 in 110*	1 in 120
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 120
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 440
Cartilage-Hair Hypoplasia (RMRP)	*1 in 500*	1 in 960
Retinoschisis, Juvenile (RS1)	*1 in 25,000*	1 in 28,000
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 590
Niemann-Pick Disease Type A & B (a.k.a. Acid Sphingomyelinase Deficiency) (SMPD1)	*1 in 160*	1 in 180
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:

Targeted mutation analysis: In solution hybridization followed by next generation sequencing at a mean read depth of >200X of the targeted mutation regions including large indels was performed on this individual's genomic DNA. If the targeted mutation region has a pseudogene, gene specific PCR and sequence analysis was performed by Sanger sequencing. 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. Please note, the c.1817_1900del84 pathogenic variant in CFTR, the 21kb deletion of exons 2 and 3 in CFTR, and the 7.6kb deletion of the promoter and exon 1 of HEXA were validated using a synthetic positive control based on published breakpoints. Due to the complexity of the sequence in the region, the following targeted mutation was unable to be analyzed in this individual: NR_003051.3(RMRP):r.71A>G. Please note, residual risk calculations may not accurately reflect this missing data.

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, NG480V09, 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.

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

■■■■■■■■■■■■
Medical Consultant
EGL Genetic Diagnostics, LLC

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Assistant Laboratory Director
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