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Name: **LAST, FIRST** MRN: **XXXXXXXX** LMM Accession ID: **PM-23-NXXXX**
DOB: **MM/DD/YYYY** Referring Facility: **HS** Specimen: **Blood, Peripheral**
Sex at birth: **Female** Referring Physician: **LAST, FIRST** Received: **MM/DD/2023**
Family #: **FXXXXXX** Copies To: **HOSPITAL SYSTEM**
Test Performed: **Expanded Genome Screening** Test Codes: **lmWGS-pnlFv4_L**

GENOME SCREENING RESULTS SUMMARY

NEGATIVE- NO MONOGENIC DISEASE FINDINGS. Sequencing of this individual's genome identified 3 carrier status variants. Pharmacogenomic associations are also included in this report. Result details are listed below.

VARIANT SUMMARY

REPORT SECTION	Disease, Inheritance	Gene Transcript	Variant	Allele State	Classification
A. MONOGENIC DISEASE FINDINGS	None identified.				
B. RISK ALLELES	None identified.				
C. CARRIER STATUS VARIANTS	Primary ciliary dyskinesia, Autosomal Recessive	CCDC40 NM_017950.4	c.248delC p.Ala83ValfsX84	Heterozygous	Pathogenic
	Primary ciliary dyskinesia, Autosomal Recessive	DNAH11 NM_001277115.2	c.12363C>G p.Tyr4121X	Heterozygous	Pathogenic
	Short/branched chain acyl-CoA dehydrogenase deficiency, Autosomal Recessive	ACADSB NM_001609.4	c.303+1G>A	Heterozygous	Pathogenic
D. PHARMACOGENOMIC ASSOCIATIONS	See below.				

DETAILED VARIANT INFORMATION

A. MONOGENIC DISEASE FINDINGS

This test did not identify any variants with the potential to cause monogenic disease in this individual.

B. RISK ALLELES

This test did not identify any reportable risk alleles (see methodology for the list of risk alleles included in this analysis).

C. CARRIER STATUS VARIANTS

This test identified 3 carrier status variants for autosomal recessive conditions. Autosomal recessive disorders are caused by the presence of pathogenic variants in both copies of the same gene. Being a carrier of these variants does not put this individual at risk for disease but may impact disease risk in this individual's children. PLEASE NOTE: The presence of a second pathogenic variant in one of these genes cannot be definitively ruled out due to the technical and analytical limitations of this assay.

FAMILIAL AND REPRODUCTIVE RISK: The risk to this individual's child or future child of developing an autosomal recessive condition is dependent on the carrier status of this individual's reproductive partner(s). Two carriers have a 25% risk for having a child with the associated disease. First degree relatives of this individual have a 50% chance of also being carriers of each of these variants. Other biologically related family members may also be carriers.

Disease, Inheritance	***Gene* Transcript**	**Variant**	**Allele State**	**Classification**
Primary ciliary dyskinesia, Autosomal Recessive	*CCDC40* NM_017950.4	c.248delC p.Ala83ValfsX84	Heterozygous	Pathogenic
Genomic Coordinate	**Location**	**Penetrance**	**Carrier Phenotype**	**Gene Coverage**
g.80039966delC (chr17, GRCh38)	Exon 3	High	None reported	100% at 15X

VARIANT INTERPRETATION: The p.Ala83ValfsX84 variant in *CCDC40* has been previously reported in 19 homozygous and 7 compound heterozygous individuals with primary ciliary dyskinesia (PCD) and segregated with disease in 1 homozygous affected relative (Becker-Heck 2011 PMID: 21131974, Nakhleh 2012 PMID: 22499950, Antony 2013 PMID: 23255504, Zariwala 2013 PMID: 23891469). This variant has been identified in 0.074% (860/1167354) of non-Finnish European chromosomes by gnomAD (http://gnomad.broadinstitute.org). However, this frequency is low enough to be consistent with a recessive allele frequency. This variant has also been reported in ClinVar (Variation ID 31069). This variant is predicted to cause a frameshift, which alters the protein's amino acid sequence beginning at position 83 and leads to a premature termination codon 84 amino acids downstream. This alteration is then predicted to lead to a truncated or absent protein. Functional studies indicate that loss of CCDC40 function results in abnormal cilia structure and motility (Becker-Heck 2011 PMID: 21131974). In summary, this variant meets criteria to be classified as pathogenic for autosomal recessive primary ciliary dyskinesia. ACMG/AMP Criteria applied: PVS1, PM3_VeryStrong, PM2_Supporting, PP1.

DISEASE INFORMATION: Primary ciliary dyskinesia is a rare genetic condition that is genetically heterogeneous. It is associated with recurrent respiratory tract infections, abnormal positioning of the internal organs, and infertility. This is caused by abnormalities in the motility of the cilia and flagella that are found in the linings of organs and tissues. Respiratory tract infections, reduced clearance of mucus, nasal congestion and chronic cough begin in early childhood and could result in bronchiectasis. Situs inversus totalis, a mirror-image reversal of all visceral organs, is found in 40-50% of individuals. Males with primary ciliary dyskinesia are frequently infertile because of abnormal sperm motility, while females with this condition are occasionally infertile likely due to abnormal cilia in the fallopian tubes. Other symptoms can include recurrent ear infections and hydrocephalus in the brain. Pathogenic variants in *CCDC40* contribute to 3-4% of primary ciliary dyskinesia (Medline Plus: https://medlineplus.gov/genetics/condition/primary-ciliary-dyskinesia, GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1122).

FAMILIAL AND REPRODUCTIVE RISK

Disease Prevalence (Estimated)	**Carrier Frequency (Estimated)**	**Reproductive Risk (Estimated)**
1/16000 (https://medlineplus.gov/genetics/condition/primary-ciliary-dyskinesia)	1/317 to 1/366 (for *CCDC40*)	1/1268 to 1/1464 (for *CCDC40*)

Disease, Inheritance	***Gene* Transcript**	**Variant**	**Allele State**	**Classification**
Primary ciliary dyskinesia, Autosomal Recessive	*DNAH11* NM_001277115.2	c.12363C>G p.Tyr4121X	Heterozygous	Pathogenic
Genomic Coordinate	**Location**	**Penetrance**	**Carrier Phenotype**	**Gene Coverage**
g.21880869C>G (chr7, GRCh38)	Exon 75	High	None reported	100% at 15X

VARIANT INTERPRETATION: The p.Tyr4121X variant in *DNAH11* has been reported in the compound heterozygous state in 1 individual with primary ciliary dyskinesia and segregated with disease in 5 affected siblings (Schwabe 2008 PMID: 18022865). It has been identified in 0.045% (29/63996) of European (Finnish) chromosomes by gnomAD (http://gnomad.broadinstitute.org, v4.0.0). However, this frequency is low enough to be consistent with the carrier frequency. This variant has also been reported in ClinVar (Variation ID 6475). This nonsense variant leads to a premature termination codon at position 4121, which is predicted to lead to a truncated or absent protein. Biallelic loss-of-function of the DNAH11 gene is an established disease mechanism in autosomal recessive primary ciliary dyskinesia 7. In summary, this variant meets criteria to be classified as pathogenic for autosomal recessive primary ciliary dyskinesia 7. ACMG/AMP Criteria applied: PVS1, PP1_Strong, PM3, PM2_Supporting.

DISEASE INFORMATION: Primary ciliary dyskinesia is a rare genetic condition that is genetically heterogeneous. It is associated with recurrent respiratory tract infections, abnormal positioning of the internal organs, and infertility. This is caused by abnormalities in the motility of the cilia and flagella that are found in the linings of organs and tissues. Respiratory tract infections, reduced clearance of mucus, nasal congestion and chronic cough begin in early childhood and could result in bronchiectasis. Situs inversus totalis, a mirror-image reversal of all visceral organs, is found in 40-50% of individuals. Males with primary ciliary dyskinesia are frequently infertile because of abnormal sperm motility, while females with this condition are occasionally infertile likely due to abnormal cilia in the fallopian tubes. Other symptoms can include recurrent ear infections and hydrocephalus in the brain. Pathogenic variants in *DNAH11* contribute to 6-9% of primary ciliary dyskinesia (Medline Plus: https://medlineplus.gov/genetics/condition/primary-ciliary-dyskinesia; GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1122).

FAMILIAL AND REPRODUCTIVE RISK

Disease Prevalence (Estimated)	**Carrier Frequency (Estimated)**	**Reproductive Risk (Estimated)**
1/16000 (https://medlineplus.gov/genetics/condition/primary-ciliary-dyskinesia)	1/211 to 1/259 (for *DNAH11*)	1/844 to 1/1036 (for *DNAH11*)

Disease, Inheritance	***Gene* Transcript**	**Variant**	**Allele State**	**Classification**
Short/branched chain acyl-CoA dehydrogenase deficiency, Autosomal Recessive	*ACADSB* NM_001609.4	c.303+1G>A	Heterozygous	Pathogenic
Genomic Coordinate	**Location**	**Penetrance**	**Carrier Phenotype**	**Gene Coverage**
g.123037848G>A (chr10, GRCh38)	Intron 3	Reduced	None reported	100% at 15X

VARIANT INTERPRETATION: The c.303+1G>A variant in *ACADSB* has been reported in the homozygous state in 2 individual with short/branched chain acyl-CoA dehydrogenase (SBCAD) deficiency; both showed the biochemical phenotype but only one showed clinical symptoms (Alfardan 2010 PMID: 20547083). It has also been reported in the homozygous state in 2 siblings without clinical symptoms (Spedicati 2021 PMID: 33727708). It has been identified in 0.056% (628/1127804) of European (non-Finnish) chromosomes, including 1 homozygote, by gnomAD (http://gnomad.broadinstitute.org, v4.0.0). This variant occurs within the canonical splice site (+/- 1,2) and is predicted to cause altered splicing leading to an abnormal or absent protein. Computational prediction tools and conservation analyses are consistent with pathogenicity. Biallelic loss-of-function of the ACADSB gene is an established disease mechanism in autosomal recessive SBCAD deficiency. SBCAD deficiency results in a biochemical phenotype that be detected from birth; however, the clinical significance of this is unclear. 90% of individuals with SBCAD deficiency have no clinical symptoms, and the remaining 10% may display developmental delay and/or neurological disorders. These 10% of individuals may represent extreme end of the clinical spectrum or coincidental findings (Porta 2019 PMID: 30730842, Alfardan 2010 PMID: 20547083). In summary, this variant meets criteria to be classified as pathogenic for autosomal recessive SBCAD deficiency. ACMG/AMP Criteria applied: PVS1, PM3_Supporting.

DISEASE INFORMATION: Short/branched chain acyl-CoA dehydrogenase deficiency (SBCAD deficiency) is a rare inborn error of metabolism that is more common among Hmong population in Southeast Asia. Affected individuals can be detected during newborn screening (NBS) but most have no clinical symptoms. 10% of SBCAD deficiency patients have developmental delay and neurological disorders; however, it is unclear if the SBCAD deficiency is causative (Porta 2019 PMID: 30730842, Medline Plus: https://medlineplus.gov/genetics/condition/short-branched-chain-acyl-coa-dehydrogenase-deficiency).

FAMILIAL AND REPRODUCTIVE RISK

Disease Prevalence (Estimated)	**Carrier Frequency (Estimated)**	**Reproductive Risk (Estimated)**
Unknown	Unknown	Unknown

D. PHARMACOGENOMIC ASSOCIATIONS

Detailed dosing instructions are not provided in the brief interpretation notes below. Extrinsic factors (e.g. diet, smoking status, co-administered medications, drug-drug interactions) and intrinsic factors (e.g. sex, age, weight, renal or hepatic function) may affect drug response. A *1/*1 or wild type result is assigned if no variants that are included in the test are detected and does not exclude the possibility that an individual has a different phenotype that may alter drug response. Patients should not use the test results to stop or change any medication unless directed by a qualified clinician. Genetic information to guide prescribing decisions may be found in a drug's FDA-approved label. These labels are found at https://www.accessdata.fda.gov/scripts/cder/daf/index.cfm and a table of current PGx biomarkers at https://www.fda.gov/medical-devices/precision-medicine/table-pharmacogenetic-associations and https://www.fda.gov/drugs/science-and-research-drugs/table-pharmacogenomic-biomarkers-drug-labeling. The Clinical Pharmacogenetics Implementation Consortium (CPIC®) website may be consulted for their most current recommendations, at https://cpicpgx.org/guidelines/; however, these have NOT received FDA-approval. Always consult a clinician or clinical pharmacologist before changing drug dosage or for additional information. The Pharmacogenomics Clinic at BWH is a resource for patients looking for a more detailed interpretation of the clinical relevance of the results (https://www.brighamandwomens.org/medicine/genetics/genetics-and-genomic-medicine/pharmacogenomics).

Genotype	**Phenotype**	**FDA Drugs with PGx Labelling**
TPMT *1/*1 NUDT15 c.415C/c.415C	TPMT Normal metabolizer NUDT15 Normal metabolizer	Azathioprine, Mercaptopurine, Thioguanine
IFNL3 c.-3180G/c.-3180G>A	IFNL3 Unfavorable response	Peginterferon Alfa-2b
CYP2C9 *1/*1	CYP2C9 Normal Metabolizer - Activity score 2	Celecoxib, Dronabinol, Erdafitinib, Flibanserin, Flurbiprofen, Lesinurad, Meloxicam, Phenytoin, Piroxicam, Siponimod
CYP2C19 *1/*17	CYP2C19 Rapid metabolizer	Brivaracetam, Carisoprodol, Citalopram, Clobazam, Clopidogrel, Dexlansoprazole, Diazepam, Doxepin, Escitalopram, Esomeprazole, Flibanserin, Omeprazole, Pantoprazole, Rabeprazole, Voriconazole

Genotype	Phenotype	FDA Drugs with PGx Labelling
CYP2C9 *1/*1 VKORC1 G/A CYP4F2 *1/*3	Use genotypes to guide dosing	Warfarin
SLCO1B1 *1A/*1A	SLCO1B1 Normal function	Elagolix, Rosuvastatin, Simvastatin
DPYD wild type at all tested positions	DPYD Normal metabolizer - Activity Score 2	Capecitabine, Fluorouracil
CYP3A5 *3/*3	CYP3A5 Poor metabolizer	Tacrolimus
UGT1A1 *1/*1	UGT1A1 Normal Metabolizer	Belinostat, Dolutegravir, Irinotecan, Nilotinib, Pazopanib, Raltegravir, Sacituzumab govitecan-hziy

RECOMMENDATIONS

These results should be interpreted in the context of this individual's personal medical history and family history. Genetic counseling is recommended for this individual and their relatives. Familial variant testing is available if desired.

COVERAGE SUMMARY

Sequencing of this individual's genome covered 97.5% of all positions at 15X coverage or higher. Please note that the presence of pathogenic variants in genes not analyzed, genes with incomplete coverage, or regions not captured by filtering strategies cannot be fully excluded.

METHODOLOGY AND LIMITATIONS

Genome sequence is generated from genomic DNA that is fragmented and barcoded. Library fragments were sequenced (2x150 base paired end) using Sequencing-By-Synthesis (SBS) chemistry and the Illumina NovaSeq sequencer with a minimum coverage of at least 20X for 90%. Sequence data were aligned to the GRCh38 assembly after discarding low quality sequences. Illumina's DRAGEN (Dynamic Read Analysis for GENomics) platform was used for demultiplexing, read mapping, genome alignment, read sorting, duplicate marking, and variant calling. Technical sensitivity of this assay is 99.10% (95% CI: 99.04-99.16%) and positive predictive value is 99.39% (95% CI: 99.37-99.41%). Variants in 4,302 genes with some level of published evidence for a gene-disease association are subsequently filtered to identify: (1) variants classified as pathogenic or likely pathogenic in our internal database; (2) variants classified as pathogenic or likely pathogenic by a ClinGen-approved expert panel; (3) variants classified as disease causing in public databases that have a minor allele frequency <5.0% in the Genome Aggregation Database (gnomAD, https://gnomad.broadinstitute.org/); and (4) nonsense, frameshift, and +/-1,2 splice-site variants in disease-associated genes with a minor allele frequency ≤0.1% in gnomAD. The evidence for phenotype-causality is then evaluated for each variant identified from the filtering strategies listed above and variants are classified based on ACMG/AMP criteria (Richards et al. 2015) with ClinGen rule specifications (https://www.clinicalgenome.org/working-groups/sequence-variant-interpretation/). Variants are reported according to HGVS nomenclature (http://varnomen.hgvs.org/). Only those variants with evidence for causing or contributing to disease are reported. All disease-associated variants on this report are confirmed via Sanger sequencing or another orthogonal technology. Please contact the laboratory for additional information.

Risk Alleles: Genotype calls for specific genomic positions are identified using the Genomic Analysis Tool Kit (GATK) and a custom script. The following likely or established risk alleles are examined and reported if identified in the "reported genotype" listed below. Some variants in these genes are associated with additional diseases and therefore other variants identified in these genes may be included on different sections of this report. Additional risk variants, if identified, may also be included on this report at the discretion of the laboratory.

Gene (Transcript)	Associated Risk	Reportable Variants	Reportable Genotypes
APC (NM_000038.4)	Colorectal Cancer	c.3920T>A (p.Ile1307Lys)	Heterozygous or homozygous
APOE (NM_000041.2)	Alzheimer's Disease	*e4 Allele*- c.388T>C (p.Cys130Arg)	Heterozygous with e2 or e3 or homozygous (e2/e4, e3/e4, e4/e4)
APOL1 (NM_003661.3)	Non-diabetic Nephropathy	*G1 Allele* – c.1164_1169delTTATAA (p.Asn388_Tyr389del) *G2 Allele* - c.[1024A>G;1152T>G] (p.[Ser342Gly;Ile384Met])	Homozygous (G1/G1 or G2/G2) or compound heterozygous (G1/G2)
CHEK2 (NM_001005735)	Breast, Colorectal, and Papillary Thyroid Cancers	c.599T>C (p.Ile200Thr)	Heterozygous or homozygous
CTRC (NM_007272.2)	Pancreatitis	c.760C>T (p.Arg254Trp)	Heterozygous or homozygous
F2 (NM_000506.3)	Venous Thromboembolism	c.*97G>A	Heterozygous or homozygous
F5 (NM_000130.4)	Factor V Deficiency	c.1601G>A (p.Arg534Gln)	Heterozygous or homozygous
GBA (NM_001005741.2)	Parkinson's Disease	c.1226A>G (p.Asn409Ser)	Heterozygous or homozygous
HFE (NM_000410.3)	Hemochromatosis	c.845G>A (p.Cys282Tyr)	Homozygous
LRRK2 (NM_198578.3)	Parkinson's Disease	c.6055G>A (p.Gly2019Ser)	Heterozygous or homozygous
MC1R (NM_002386)	Melanoma	c.880G>C (p.Asp294His)	Heterozygous or homozygous
MITF (NM_000248.3)	Melanoma	c.952G>A (p.Glu318Lys)	Heterozygous or homozygous
MUC5B (NM_002458.2)	Pulmonary Fibrosis	c.-3133G>T	Heterozygous or homozygous
PNPLA3 (NM_025225.2)	Non-alcoholic Fatty Liver Disease Type 1	c.444C>G (p.Ile148Met)	Homozygous
PRNP (NM_000311.3)	Prion Disease	c.628G>A (p.Val210Ile)	Heterozygous or homozygous

SERPINA1 (NM_001127701.1)	Alpha-1 Antitrypsin Deficiency	*S Allele*- c.863A>T (p.Glu288Val) *Z Allele*- c.1096G>A (p.Glu366Lys)	Homozygous Z allele (Z/Z) or compound heterozygous Z and S allele (Z/S)
SERPINC1 (NM_000488)	Venous Thromboembolism	c.1246G>T (p.Ala416Ser)	Heterozygous or homozygous
SPINK1 (NM_003122)	Pancreatitis	c.101A>G (p.Asn34Ser)	Heterozygous or homozygous

PGx: Genotype calls for specific genomic positions are identified using the Genomic Analysis Tool Kit (GATK) and a custom script. Diplotypes and phenotypes are generated using the Clinical Pharmacogenetics Implementation Consortium (CPIC®) allele tables (https://cpicpgx.org/guidelines/) and custom scripts. The following pharmacogenomic variants are detected by this assay: *CYP2C19* (NM_000769.1): **2* (rs4244285/c.681G>A, rs12769205/c.332-23A>G), **3* (rs4986893/c.636G>A), **4A* (rs28399504/c.1A>G), **4B* (rs28399504/c.1A>G, rs12248560/c.-806C>T), **5* (rs56337013/c.1297C>T), **6* (rs72552267/c.395G>A), **7* (rs72558186/c.819+2T>A), **8* (rs41291556/c.358T>C), **17* (rs12248560/c.-806C>T), **35* (rs12769205/c.332-23A>G); *CYP2C9* (NM_000771.3): **2* (rs1799853/c.430C>T), **3* (rs1057910/c.1075A>C), **5* (rs28371686/c.1080C>G), **6* (rs9332131/c.818del), **8* (rs7900194/c.449G>A), **11* (rs28371685/c.1003C>T), **13* (rs72558187/c.269T>C); *CYP3A5* (NM_000777.4): **3* (rs776746/c.219-237A>G), **6* (rs10264272/c.624G>A), **7* (rs41303343/c.1035dup); *CYP4F2* (NM_001082.4): **3* (rs2108622/c.1297G>A); *DPYD* (NM_000110.3): rs3918290/c.1905+1G>A, rs55886062/c.1679T>G, rs67376798/c.2846A>T, rs72549309/c.299_302del/legacy name c.295_299TCATT>T, rs115232898/c.557A>G, rs1801266/c.703C>T, rs78060119/c.1156G>T, rs56038477/c.1236G>A, rs72549303/c.1898del, rs1801268/c.2983G>T, rs75017182/c.1129-5923C>G; *IFNL3* (NM_172139.3): rs12979860/c.-3180G>A; *NUDT15* (NM_018283.3): rs116855232/c.415C>T; *SLCO1B1* (NM_006446.4): rs4149056/c.521T>C; *TPMT* (NM_000367.4): **2* (rs1800462/c.238G>C), **3A* (rs1800460/c.460G>A, rs1142345/c.719A>G), **3B* (rs1800460/c.460G>A), **3C* (rs1142345/c.719A>G), **4* (rs1800584/c.626-1G>A); *VKORC1* (NM_024006.4): rs9923231/c.-1639G>A; *UGT1A1* (NM_000463.3): **6* (rs4148323/c.211G>A), **27* (rs35350960/c.686C>A), **36* (rs3064744/c.-53TA[6]), **28* (rs3064744/ c.-53TA[8]), **37* (rs3064744/ c.-53TA[9]). Variant and star (*) allele definitions are available for download at https://personalizedmedicine.partners.org/Laboratory-For-Molecular-Medicine/Tests/Genomic%20Screening%20Spray.aspx (see the link "details here" in the Methodology section of this webpage). Additionally, variants in *G6PD*, *RYR1*, and *CACNA1S* associated with an altered metabolism status are reported, if identified. Detected variants include those that are clinically relevant based on allele function and frequency information found in tables created by PharmGKB and CPIC (https://www.pharmgkb.org/page/pgxGeneRef). This test does not report all pharmacogenomic variants that might alter protein function. A *1/*1 or wild type result is assigned if no variants that are included in the test are detected. Therefore, a *1/*1 or other result does not exclude the possibility that an individual has a different phenotype that may alter drug response. This risk may vary among ancestries. This assay cannot determine if multiple variants in the same gene are present in cis or trans, leading to an inability to definitively assign a diplotype and phenotype. This test does not detect copy number variants. Genetic information to guide prescribing decisions may be found in a drug's FDA-approved label. These labels are found at https://www.accessdata.fda.gov/scripts/cder/daf/index.cfm and a table of current PGx biomarkers at https://www.fda.gov/medical-devices/precision-medicine/table-pharmacogenetic-associations and https://www.fda.gov/drugs/science-and-research-drugs/table-pharmacogenomic-biomarkers-drug-labeling.

Limitations: Specific types of genetic variation, such as short tandem repeats (including triplet repeat expansions), structural variation, and large copy number events are currently not reliably detected by this assay. Additionally, while genome sequencing covers ~95% of the genome; there are certain regions for which the assay may fail to adequately generate sequence information, such as regions of high homology or regions that are highly repetitive as well as regions with low coverage, mapping quality or base quality. Moreover, not all disease-associated genes have been identified and the clinical significance of variation in many genes is not well understood. The sequencing data is limited to the cell-type (material) source used for DNA extraction. Additionally, not all variants identified have been analyzed. Variant interpretation may change over time if more information becomes available.

The initial sequencing component of this test was performed by the Clinical Research Sequencing Platform of the Broad Institute (320 Charles St, Cambridge, MA 02141; CLIA#22D2055652), and the Sanger confirmation, interpretive algorithms and clinical reports were generated by the Laboratory for Molecular Medicine at Partners Healthcare Personalized Medicine (LMM, 65 Landsdowne St, Cambridge, MA 02139; 617-768-8500; CLIA#22D1005307). This test 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.

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