Gene Tox Lab Solutions

Patient Information	Specimen Details	Provider Information
Name: Jane Jane Jane Doedoe Doedoe Doedoe	**Specimen Type:** Blood	**Physician:** You know who
Acc#: 20180514SC - Birgit	**Collection Date:** 12/31/2018	**Report Date:** Not yet approved
DOB: 12/26/1995	**Received Date:** 12/31/2018	
Sex: Female		

Preventive Care - ACMG 59 Report

Indication for Testing

Family history of some cancer

Test Result

Positive Result Pathogenic/Likely pathogenic variant(s) detected.

Test Details

GENE	VARIANT	POSITION	ZYGOSITY	CLASSIFICATION
LMNA	c.908_909delCT p.Ser303CysfsTer27	g.156105070CCT>C	Homozygous	Pathogenic
SDHC	c.78-1G>A	g.161298185G>A	Homozygous	Likely pathogenic

Summary

***LMNA* p.Ser303CysfsTer27**

This patient is homozygous for variant p.Ser303CysfsTer27 in the *LMNA* gene.

The nuclear lamina consists of a two-dimensional matrix of proteins located next to the inner nuclear membrane. The lamin family of proteins make up the matrix and are highly conserved in evolution. During mitosis, the lamina matrix is reversibly disassembled as the lamin proteins are phosphorylated. Lamin proteins are thought to be involved in nuclear stability, chromatin structure and gene expression. Vertebrate lamins consist of two types, A and B. Alternative splicing results in multiple transcript variants. Mutations in this gene lead to several diseases: Emery-Dreifuss muscular dystrophy, familial partial lipodystrophy, limb girdle muscular dystrophy, dilated cardiomyopathy, Charcot-Marie-Tooth disease, and Hutchinson-Gilford progeria syndrome. [provided by RefSeq, Apr 2012]

***SDHC* c.78-1G>A**

This patient is homozygous for variant c.78-1G>A associated with the *SDHC* gene.

This gene encodes one of four nuclear-encoded subunits that comprise succinate dehydrogenase, also known as mitochondrial complex II, a key enzyme complex of the tricarboxylic acid cycle and aerobic respiratory chains of mitochondria. The encoded protein is one of two integral membrane proteins that anchor other subunits of the complex, which form the catalytic core, to the inner mitochondrial membrane. There are several related pseudogenes for this gene on different chromosomes. Mutations in this gene have been associated with paragangliomas. Alternatively spliced transcript variants have been described. [provided by RefSeq, May 2013]

Recommendations

- The findings of this report should be considered as educational to the patient and the clinician.
- The findings should not be interpreted as a medical diagnosis, advice, or consultation from a professional healthcare provider.
- In an event of a Positive Result (Pathogenic/Likely Pathogenic variant detected), patients are advised to refer to physicians for proper follow-up examination.

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Disclaimer

Limitations: This test will not detect all the known alleles that result in altered or inactive tested genes. This test does not account for all individual variations in the individual tested. Absence of a detectable gene mutation does not rule out the possibility that a patient has different phenotypes due to the presence of an undetected polymorphism or due to other factors such as drug-drug interactions, comorbidities, and lifestyle habits.

Methodology: Single-nucleotide polymorphism (SNP)-based assays detect listed alleles, including all common and most rare variants with known clinical significance at analytical sensitivity and specificity >99%. Genotyping analysis was performed on Illumina Global Screening Array, which covers over 710,000 SNPs. 15,296 SNP information located in ACMG 59 genes were used for this test.

Signature will be placed here once report is approved

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About the Report

This report offers insights on pathogenic/likely pathogenic variants found within the set of 59 core health genes as recommended by the American College of Medical Genetics and Genomics (ACMG). These variants were elucidated from extracted genomic material analyzed on an Illumina Global Screening Array-based assay designed to detect clinically relevant single-nucleotide polymorphisms (SNPs). Detected variants were evaluated using genotype quality, minor allele frequency, and curated clinical database for pathogenicity. The genes included in this report adhere to ACMG's secondary findings recommendations, most recently updated on November 2016.

Genes tested and related diseases

ACTA2	Marfan syndrome, Loeys-Dietz syndromes, and familial thoracic aortic aneurysms and dissections
ACTC1	Hypertrophic cardiomyopathy, dilated cardomyopathy
APC	Familial adenomatous polyposis
APOB	Familial hypercholesterolemia
ATP7B	Wilson disease
BMPR1A	Juvenile polyposis
BRCA1	Hereditary breast and ovarian cancer
BRCA2	Hereditary breast and ovarian cancer
CACNA1S	Malignant hyperthermia susceptibility
COL3A1	Ehlers-Danlos syndrome, vascular type
DSC2	Arrhythmogenic right venticular cardiomyopathy
DSG2	Arrhythmogenic right venticular cardiomyopathy
DSP	Arrhythmogenic right venticular cardiomyopathy
FBN1	Marfan syndrome, Loeys-Dietz syndromes, and familial thoracic aortic aneurysms and dissections
GLA	Hypertrophic cardiomyopathy, dilated cardomyopathy
KCNH2	Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome
KCNQ1	Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome
LDLR	Familial hypercholesterolemia
LMNA	Hypertrophic cardiomyopathy, dilated cardomyopathy
MEN1	Multiple endocrine neoplasia type 1
MLH1	Lynch syndrome
MSH2	Lynch syndrome
MSH6	Lynch syndrome
MUTYH	MYH-associated polyposis; adenomas, multiple colorectal, FAP type 2; colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas
MYBPC3	Hypertrophic cardiomyopathy, dilated cardomyopathy
MYH11	Marfan syndrome, Loeys-Dietz syndromes, and familial thoracic aortic aneurysms and dissections
MYH7	Hypertrophic cardiomyopathy, dilated cardomyopathy
MYL2	Hypertrophic cardiomyopathy, dilated cardomyopathy
MYL3	Hypertrophic cardiomyopathy, dilated cardomyopathy
NF2	Neurofibromatosis type 2
OTC	Ornithine transcarbamylase deficiency
PCSK9	Familial hypercholesterolemia
PKP2	Arrhythmogenic right venticular cardiomyopathy
PMS2	Lynch syndrome
PRKAG2	Hypertrophic cardiomyopathy, dilated cardomyopathy
PTEN	PTEN hamartoma tumor syndrome
RB1	Retinoblastoma
RET	Familial medullary thyriod cancer, multiple endocrine neoplasia type 2
RYR1	Malignant hyperthermia susceptibility
RYR2	Catecholaminergic polymorphic ventricular tachycardia
SCN5A	Romano-Ward long-QT syndrome types 1, 2, and 3; Brugada syndrome
SDHAF2	Hereditary paraganglioma-phechromocytoma syndrome
SDHB	Hereditary paraganglioma-phechromocytoma syndrome
SDHC	Hereditary paraganglioma-phechromocytoma syndrome
SDHD	Hereditary paraganglioma-phechromocytoma syndrome
SMAD3	Marfan syndrome, Loeys-Dietz syndromes, and familial thoracic aortic aneurysms and dissections
SMAD4	Juvenile polyposis
STK11	Peutz-Jeghers syndrome
TGFBR1	Marfan syndrome, Loeys-Dietz syndromes, and familial thoracic aortic aneurysms and dissections
TGFBR2	Marfan syndrome, Loeys-Dietz syndromes, and familial thoracic aortic aneurysms and dissections
TMEM43	Arrhythmogenic right venticular cardiomyopathy
TNNI3	Hypertrophic cardiomyopathy, dilated cardomyopathy
TNNT2	Hypertrophic cardiomyopathy, dilated cardomyopathy
TP53	Li-Fraumeri synfrime
TPM1	Hypertrophic cardiomyopathy, dilated cardomyopathy
TSC1	Tuberous sclerosis complex
TSC2	Tuberous sclerosis complex
VHL	Von Hippel-Lindau syndrome
WT1	WT1-related Wilms tumor