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Client: ARUP Physician Services
321 TESTING ANSR EXTRACT
Salt Lake City, NY 84108
UNITED STATES

Physician: TEST,

Patient: PROD, TRACKEXADD NO CONSENT

DOB	5/17/1994
Gender:	Male
Patient Identifiers:	546194
Visit Number (FIN):	569124
Collection Date:	5/17/2019 10:24

Exome Sequencing, Familial Control

ARUP test code 2006340

Exome Sequencing, Familial Control	See Note

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Control sample: No consent signed
This individual, who is serving as a control for exome sequencing for another family member, did not sign a consent form; thus, any incidental findings from exome sequencing will not be analyzed or resulted.

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., Ph.D.

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: Exome Sequencing, Familial Control

CHARACTERISTICS: DNA coding regions and intron/exon boundaries of the human exome are sequenced to identify the cause(s) of a disorder in a family member. The American College of Medical Genetics (ACMG) recommends analysis of the following genes for pathogenic mutations in all individuals undergoing exome sequencing:

GENES ASSOCIATED WITH AN INCREASED RISK FOR TUMORS/CANCER: hereditary breast and ovarian cancer (BRCA1, BRCA2), Li-Fraumeni syndrome (TP53), Peutz-Jeghers syndrome (STK11), Lynch syndrome (MLH1, MSH2, MSH6, PMS2), familial adenomatous polyposis (APC), MUTYH-associated polyposis, Von Hippel Lindau syndrome (VHL), multiple endocrine neoplasia type 1 (MEN1), multiple endocrine neoplasia type 2/familial medullary thyroid cancer (RET), PTEN hamartoma tumor syndrome (PTEN), retinoblastoma (RB1), hereditary paraganglioma-pheochromocytoma syndrome (SDHD, SDHAF2, SDHC, SDHB), tuberous sclerosis complex (TSC1, TSC2), WT1-related Wilms (WT1), neurofibromatosis type 2 (NF2). GENES ASSOCIATED WITH CARDIOVASCULAR (HEART) PROBLEMS: EDS IV (COL3A1), Marfan syndrome (FBN1), Loeys-Dietz syndrome (TGFBR1 and TGFBR2), familial thoracic aortic aneurysms and dissections (SMAD3, ACTA2, MYLK, MYH11), hypertrophic cardiomyopathy/dilated cardiomyopathy (MYBPC3, MYH7, TNNT2, TNNI3, TPM1, MYL3, ACTC1, PRKAG2, GLA ,MYL2, LMNA), catecholaminergic polymorphic ventricular tachycardia (RYR2), arrhythmogenic right ventricular cardiomyopathy (PKP2, DSP, DSC2, TMEM43, DSG2), Romano-Ward long QT syndromes types 1, 2, and 3, Brugada syndrome (KCNQ1, KCNH2, SCN5A), familial hypercholesterolemia (LDLR, APOB, PCSK9).

GENES INFLUENCING RESPONSE TO ANESTHESIA: malignant hyperthermia (RYR1, CACNA1S).

INHERITANCE: Varies depending on the specific gene and variant.

CLINICAL SENSITIVITY: Varies by gene.

METHODOLOGY: Targeted capture of all coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to confirm reported variants. Human genome build 19 (Hg 19) was used for data analysis.

LIMITATIONS OF ANALYSIS: Not all pathogenic variants occur in the coding regions of genes. Some genes, or parts of genes, may not be adequately sequenced to allow for confident analysis. The following types of variants may not be detectable: those located in genes with corresponding pseudogenes, those in repetitive or high GC rich regions, large deletions / duplications / rearrangements, and mosaic mutations. Rare variants in probe hybridization sites may compromise analytical sensitivity. Mode of inheritance, reduced penetrance,and genetic heterogeneity could reduce the clinical sensitivity.

LIMITATIONS OF REPORTING: Only known pathogenic variants identified in genes on the ACMG-recommended panel are reported. Variants of unknown significance will not be reported. Single pathogenic variants in autosomal recessive genes will not be reported.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Exome Sequencing, Familial Control	19-137-104882	5/17/2019 10:24:00 AM	5/17/2019 10:25:23 AM	5/17/2019 1:36:00 PM

END OF CHART

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