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Patient Report

Patient Age/Gender: 39 years Male

Specimen Collected: 23-Jun-21 16:45

Loeys-Dietz Syndrome Core Panel by NGS **|Received: 23-Jun-21 16:45** **Report/Verified: 23-Jun-21 16:46**

Procedure	Result	Units	Reference Interval
Spcm LDS	Whole Blood		
LDS Interp	Positive [f1 i1]		

Result Footnote

f1: LDS Interp

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.

Test Information

i1: LDS Interp

BACKGROUND INFORMATION: Loeys-Dietz Syndrome Core Panel, Sequencing

CHARACTERISTICS: Cardiovascular findings (aortic dissection, arterial aneurysms, arterial tortuosity, MVP), skeletal abnormalities (arachnodactyly, talipes equinovarus, joint laxity, cervical spine malformations and instability, pectus excavatum and carinatum), craniofacial features (hypertelorism, retrognathia, craniosynostosis, and bifid uvula), cutaneous findings (translucent velvety skin, visible veins in chest, widened poorly-formed scars, and easy bruising), allergy and gastrointestinal disease (asthma, allergic rhinitis, food allergy, eosinophilic gastrointestinal disease) and spontaneous rupture of spleen, bowel, and uterus during pregnancy).

EPIDEMIOLOGY: Unknown.

CAUSE: Pathogenic germline variants in SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, and TGFBR2.

INHERITANCE: Autosomal dominant; 75 percent of cases are caused by a de novo variant.

PENETRANCE: High

CLINICAL SENSITIVITY: Approximately 75-85 percent.

GENES TESTED: TGFBR1, TGFBR2.

METHODOLOGY: 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 fill in regions of low coverage and confirm reported variants.

Test Information

i1: LDS Interp

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity of this test is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions from 1-10 base pairs in size. Variants greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced.

LIMITATIONS: A negative result does not exclude a diagnosis of Loeys-Dietz syndrome. This test only detects variants within the coding regions and intron-exon boundaries of the TGFBR1 and TGFBR2 genes. Variants in other genes, causing LDS (SMAD2, SMAD3, TGFB2, TGFB3) are not analyzed by this core panel. Regulatory region variants and deep intronic variants will not be identified. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. This assay may not detect low-level mosaic or somatic variants associated with disease. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Noncoding transcripts were not analyzed.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.