ARUP LABORATORIES | aruplab.com
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Tracy I. George, MD, Chief Medical Officer

Client: ARUP Physician Services
321 TESTING ANSR EXTRACT
Salt Lake City, NY 84108-
USA

Provider: 68912 -arup,arup

Patient: **PBD NGS, SAMPLE6**
DOB:
Gender: Female
Patient Identifiers: 599179
Visit Number (FIN): 623251
Client Supplied ID:

Specimen Collected: 25-Aug-21 12:57

Peroxisomal Disorders Panel by NGS | **Received: 26-Aug-21 12:02** **Report/Verified: 26-Aug-21 12:18**

Procedure	Result	Units	Reference Interval
Peroxisomal Disorders Specimen	Whole Blood		
Peroxisomal Disorders Interp	See Note [f1] [i1]		

Result Footnote

f1: Peroxisomal Disorders 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: Peroxisomal Disorders Interp

BACKGROUND INFORMATION: Peroxisomal Disorders Panel, Sequencing

CHARACTERISTICS: Peroxisomal disorders are a group of diseases caused by gene defects impairing the formation (peroxisome biogenesis disorders) or function of the peroxisomes, with symptoms that impact a wide range of body systems. Peroxisome biogenesis disorders include Zellweger spectrum disorders (ZSD) and rhizomelic chondrodysplasia punctata (RCDP). Single enzyme defects include Refsum disease, peroxisomal acyl-CoA oxidase deficiency, peroxisomal bifunctional deficiency, defects of bile acid synthesis, and primary hyperoxaluria. Some single enzyme defects present with similar clinical features to ZSD (e.g. ACOX1, HSD17B4) or RCDP (e.g. AGPS, GNPAT), although these often can be distinguished by extensive biochemical testing. Signs and symptoms of peroxisomal disorders may develop as early as the newborn period, with hypotonia, seizures, poor growth, and feeding problems. Leukodystrophy, hepatic dysfunction, adrenal insufficiency, hearing loss, and visual impairment may also be present. Skeletal abnormalities in individuals with peroxisomal disorders include stippling of the growth plates and chondrodysplasia punctata, or progressive loss of bone mineral density. Later onset forms of these conditions have similar symptoms, but with a slower progression and milder severity. Developmental delay and intellectual disability are common.

INCIDENCE: Approximately 1 in 50,000

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

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CAUSE: Pathogenic germline variants in genes associated with the structure and function of peroxisomes.

INHERITANCE: Autosomal recessive with rare autosomal dominant cases

CLINICAL SENSITIVITY: At least 97% for Zellweger spectrum disorders
At least 97% for rhizomelic chondrodysplasia punctata

GENES TESTED: ABCD3, ACBD5*, ACOX1, AGPS, AGXT, AMACR, DNM1L, FAR1, GNPAT, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, SCP2*

* One or more exons are not covered by sequencing for the indicated gene; see limitations section below.

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.

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 peroxisomal disorders. This test only detects variants within the coding regions and intron-exon boundaries of the targeted genes. 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. Non coding transcripts were not analyzed.

The following regions are not sequenced due to technical limitations of the assay:

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ACBD5 (NM_001352568) exon(s) 6
ACBD5 (NM_001352569) exon(s) 6
ACBD5 (NM_001352570) exon(s) 13
ACBD5 (NM_001352571) exon(s) 5
ACBD5 (NM_001352573) exon(s) 6
ACBD5 (NM_001352574) exon(s) 6
ACBD5 (NM_001352575) exon(s) 6
ACBD5 (NM_001352576) exon(s) 6
ACBD5 (NM_001352581) exon(s) 6
ACBD5 (NM_001352585) exon(s) 5
ACBD5 (NM_001352586) exon(s) 5
ACBD5 (NM_001352568) partial exon(s) 1(Chr10:27529638-27529648)
ACBD5 (NM_001352572) partial exon(s) 1(Chr10:27529638-27529648)
SCP2 (NM_001007098) exon(s) 11
SCP2 (NM_001330587) exon(s) 12

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.