

Client: Example Client ABC123 123 Test Drive Salt Lake City, UT 84108 UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB 1/26/1997 **Gender:** Male

Patient Identifiers: 01234567890ABCD, 012345

Visit Number (FIN): 01234567890ABCD **Collection Date:** 00/00/0000 00:00

TPSAB1 Copy Number Analysis by ddPCR

ARUP test code 3017399

TPSAB1, Source Whole Blood

Alpha-tryptase Copy Number 2 Copies

Beta-tryptase Copy Number 2 Copies

TPSAB1 Interp Not Increased

There is no increase in copy number of TPSAB1 (alpha-tryptase). Calculations are based on the allelic ratio of TPSAB1 to AP3B1, and TPSB2 to AP3B1 genes.

This result has been reviewed and approved by

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

4848



INTERPRETIVE INFORMATION: TPSAB1 Copy Number Analysis by ddPCR

INHERITANCE: autosomal dominant CAUSE: increased copy number of TPSAB1 gene on a single allele VARIANTS TESTED: copy number of TPSAB1 and TPSB2 genes. Methodology: extracted DNA from whole blood or bone marrow specimens is amplified in a droplet digital polymerase chain reaction (ddPCR) targeting the TPSAB1, TPSB2, along with a reference gene AP3B1. Three allele-specific hydrolysis probes are used for the detection. Results are reported as integer copy numbers.

ANALYTICAL SENSITIVITY AND SPECIFICITY: greater than 99 percent ANALYTICAL SENSITIVITY AND SPECIFICITY: greater than 99 percent CLINICAL SENSITIVITY: greater than 99 percent LIMITATIONS: Diagnostic errors may occur due to rare sequence and copy number variations. Single base pair substitutions, small deletions/duplications, and regulatory regions are not detected. This test is unable to determine chromosomal phase of TPSAB1 and TPSB2 genes. This assay detects only total number of TPSAB1 and TPSB2 gene copies. Therefore, rare copy number changes that affect TPSAB1 allelic/chromosomal distribution are not detected by this assay. The results of this test must always be interpreted in the

The results of this test must always be interpreted in the context of other relevant clinical data. Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
TPSAB1, Source	24-233-401608	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-tryptase Copy Number	24-233-401608	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Beta-tryptase Copy Number	24-233-401608	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
TPSAB1 Interp	24-233-401608	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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

Patient: Patient, Example ARUP Accession: 24-233-401608 Patient Identifiers: 01234567890ABCD, 012345 Visit Number (FIN): 01234567890ABCD Page 2 of 2 | Printed: 9/4/2024 2:07:44 PM

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