

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 7/4/1966  
**Gender:** Female  
**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 3 Copies

TPSAB1 Interp **Increased \***

There is an increase in copy number of TPSAB1 (alpha-tryptase), which is reported in hereditary alpha tryptasemia (HaT). Calculations are based on the allelic ratio of TPSAB1 to AP3B1, and TPSB2 to AP3B1 genes.

This result has been reviewed and approved by [REDACTED]

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

Unless otherwise indicated, testing performed at:

**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  
**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 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	24-240-401676	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha-tryptase Copy Number	24-240-401676	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Beta-tryptase Copy Number	24-240-401676	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
TPSAB1 Interp	24-240-401676	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**

Unless otherwise indicated, testing performed at: