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PATIENT REPORT

Patient Age/Sex: 60 years Male

Specimen Collected: 13-Mar-24 13:29

TPSAB1 Copy Number Analysis by ddPCR | **Received: 13-Mar-24 13:30** **Report/Verified: 14-Mar-24 09:44**

Procedure	Result	Units	Reference Interval
TPSAB1,Source	Whole Blood		
Alpha-tryptase Copy Number	2 copies		
Beta-tryptase Copy Number	3 copies		
TPSAB1 Interp	**Increased** * f1 i1		

Result Footnote

f1: TPSAB1 Interp

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 Ganna Shestakova, M.D., Ph.D.

Test Information

i1: TPSAB1 Interp

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.