VIRANT DIAGNOSTICS

CLIA Certified and CAP Accredited Laboratory

Alpha-tryptasemia CNV Result Interpretation

For Healthcare Providers

The **Alpha-tryptasemia Copy Number Variation** test reports the number of α-tryptase and β-tryptase copies encoded by the TPSAB1 gene. There are many different possible genotypes. A genotype is either normal or abnormal.

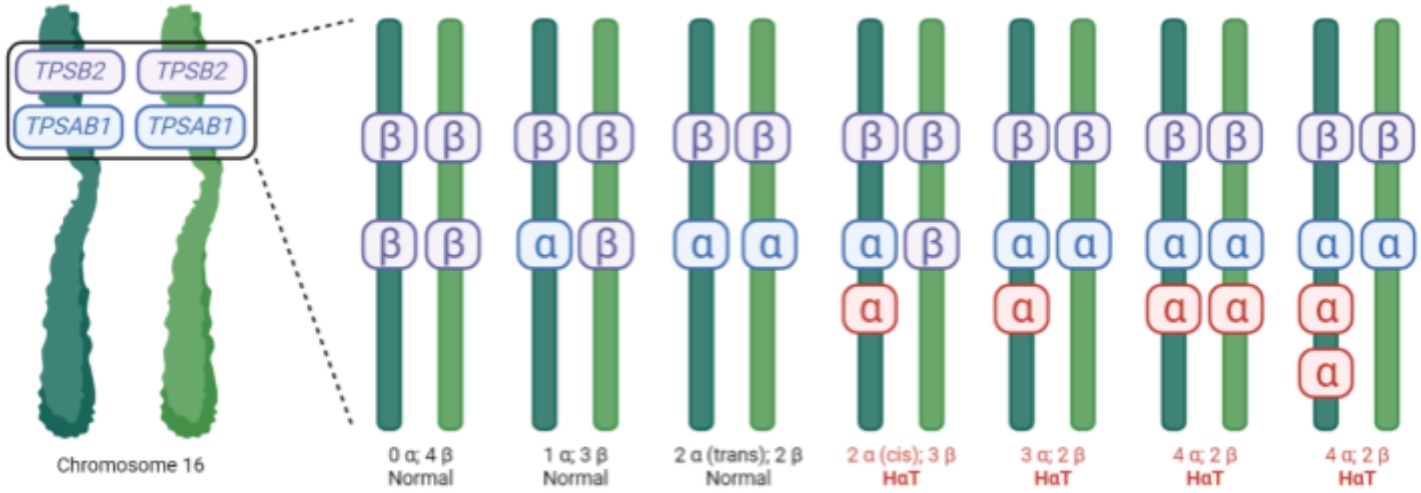

Image created using BioRender

Your patient's results: **Normal**	*The patient does not have the hereditary alpha-tryptasemia trait.*
Meaning:	No more than 2 copies of α-tryptase on opposite alleles (trans)
Clinical Significance:	Patients with a negative report but high clinical suspicion should be monitored closely.

Your patient's results: **Abnormal**	*The patient has the hereditary alpha-tryptasemia trait.*
Meaning:	3 or more total copies of α-tryptase OR 2 or more copies of α-tryptase on the same allele (cis)
Clinical Significance:	**Hereditary alpha-tryptasemia (HαT)** is when a patient has additional copies of the ***TPSAB1* gene**. The *TPSAB1* gene encodes both **α-tryptase** and **β-tryptase**, which encode for the mast cell mediator **tryptase**. Additional copies of α-tryptase lead to elevated blood tryptase levels (it is unclear what additional copies of β-tryptase mean and is considered benign). Increased tryptase levels cause HαT symptoms. Increased copies of α-tryptase are predictive of higher blood tryptase levels (i.e., a patient with 4 α-tryptase copies would have greater disease severity than a patient with 3 α-tryptase copies). Even if a patient carries the trait for hereditary alpha-tryptasemia, they may not experience symptoms; however, they are still capable of passing down the trait and having a symptomatic child.

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