VIRANT DIAGNOSTICS

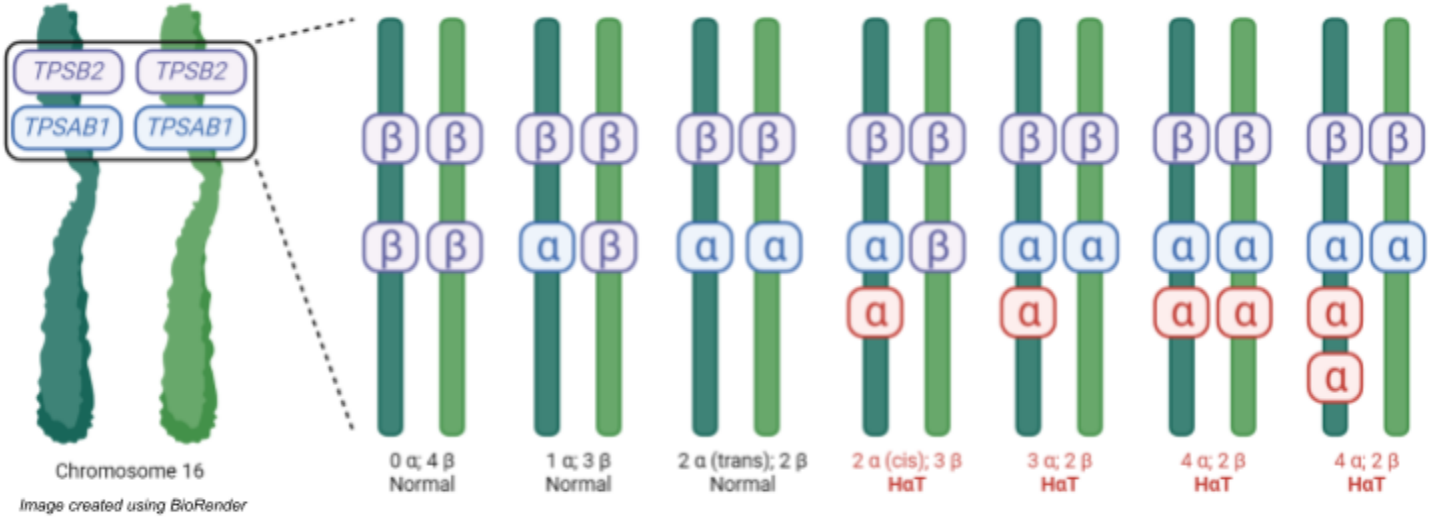
CLIA-Certified & CAP-Accredited Laboratory

MCD GENETIC TESTING RESULT INTERPRETATION

For Healthcare Providers

Alpha-tryptasemia Copy Number Variation Result Interpretation

Your patient's result	Meaning	Clinical Significance
Abnormal	3 or more total copies of α-tryptase OR 2 or more copies of α-tryptase on the same allele (cis)	**The patient has the hereditary alpha-tryptasemia trait.** **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.
Normal	No more than 2 copies of α-tryptase on opposite alleles (trans)	**The patient does not have the hereditary alpha-tryptasemia trait.** Patients with a negative report but high clinical suspicion of hereditary alpha-tryptasemia should be monitored closely.



High Sensitivity *KIT* D816V Mutation Hotspot Result Interpretation

Your patient's result	Meaning	Clinical Significance
Positive	*KIT* D816V mutated allele percentage is ≥ 0.015%	**The patient satisfies a minor criterion for the diagnosis of systemic mastocytosis.** **Systemic mastocytosis (SM)** is when a patient has abnormal proliferation and accumulation of mast cells in multiple, noncutaneous organ systems. The ***KIT* gene** produces a **receptor tyrosine kinase** that controls the growth and development of mast cells. A specific mutation in the *KIT* gene, *KIT* c.2447A>T p.D816V, known as ***KIT* D816V**, produces a dysfunctional form of this receptor that causes increased mast cell growth, development, and mediator release. Increased levels of mast cell mediators, such as **histamine** and **tryptase**, cause SM symptoms. Detection of the *KIT* D816V mutation satisfies a minor criterion for the diagnosis of systemic mastocytosis. A higher percentage of mutated alleles indicates increased risk for disease. The *KIT* D816V mutation is a somatic mutation and cannot be passed down.
Negative	*KIT* D816V mutated allele percentage is 0%	**The patient does not satisfy a minor criterion for the diagnosis of systemic mastocytosis.** Patients with a negative report but high clinical suspicion of systemic mastocytosis should be monitored closely with consideration of repeat testing if their condition worsens.
Inconclusive	*KIT* D816V mutated allele percentage is 0.005-0.015%	**Repeat testing is necessary to determine if the patient satisfies a minor criterion for the diagnosis of systemic mastocytosis.** The presence of the *KIT* D816V mutation cannot be confirmed and repeat testing should be performed with a new sample.

Virant Diagnostics, Inc.
11002 Veirs Mill Rd, #404, Wheaton, MD 20902
Website: virantdx.com Email: info@virantdx.com
Phone: 877-888-2973 Fax: 888-713-3456