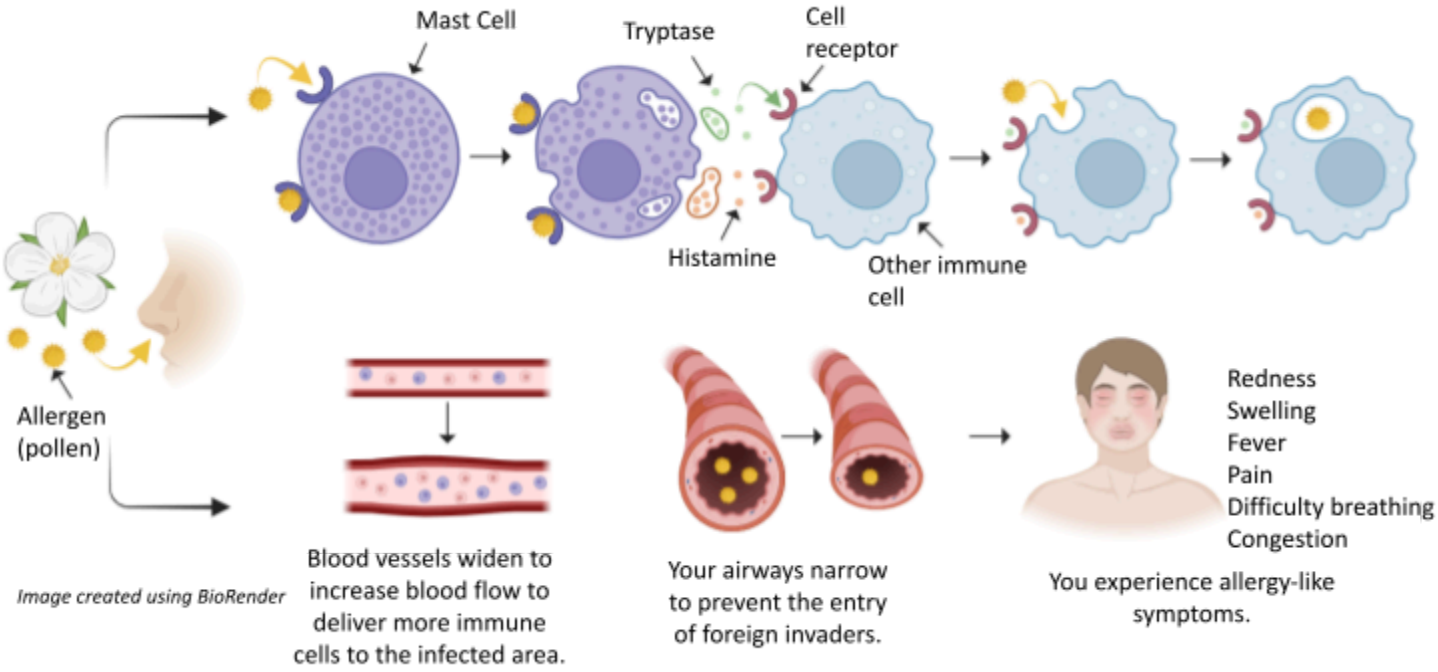
VIRANT DIAGNOSTICS

CLIA Certified and CAP Accredited Laboratory

ABOUT MAST CELL DISORDERS

For Patients

Your **immune system** protects your body from infection with the help of various, specialized **immune cells**. **Mast cells** are a special type of immune cell that are located throughout your body in areas that interact with the outside world, such as in your skin, lungs, and stomach. When they recognize a foreign invader, such as an allergen or virus, they release chemicals, known as **mediators**; two important mast cell mediators are **histamine** and **tryptase**. These mediators bind to **receptors** on other immune cells to trigger them to attack the foreign invader. This is one way your immune system protects your body and keeps you healthy.



Mast cell disorders (MCDs) occur when your body produces too many mast cells and/or your mast cells are overreacting. This leads to an increase in the release of mast cell mediators. Increased levels of mast cell mediators cause uncomfortable symptoms of an inflammatory response. So, patients with MCDs often experience recurring, severe allergy-like symptoms and may even experience anaphylaxis. MCDs include hereditary alpha-tryptasemia (HαT), systemic mastocytosis (SM), cutaneous mastocytosis (CM), and mast cell activation syndrome (MCAS).

MCD **treatment** focuses on reducing the frequency and severity of attacks:

Avoid known triggers

- Extreme heat/cold
- Sunlight
- Strong smells
- Certain foods and beverages, particularly spicy foods and alcohol
- Social, emotional, and physical (exercise, fatigue) stress
- Insect stings
- Skin irritation/friction (e.g., massage)
- Certain medications (e.g., antibiotics, opioids, aspirin)
- Pathogens (e.g., mold, bacteria, viruses)

Medications

- Antihistamines (e.g., Benadryl®): block histamine activity
- Mast cell stabilizers: control mast cell activity
- Leukotriene inhibitors (e.g., Singulair®): block leukotriene (mast cell mediator) activity
- Aspirin therapy (under the supervision of a healthcare provider)
- Anti-IgE therapy
- Chemotherapy, including *KIT* D816V therapy when the mutation is detected

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Hereditary Alpha-tryptasemia (HαT)	
Description & Cause	**Hereditary alpha-tryptasemia** (HαT) is a genetic condition with additional copies of a gene called TPSAB1 that produces tryptase. Additional copies of the *TPSAB1* gene lead to elevated tryptase levels, causing inflammatory responses and HαT symptoms.
Symptoms	• Anaphylaxis • Skin flushing, itching, hives, swelling • Palpitations, changes in blood pressure, dizziness, fainting • Abdominal pain, bloating, diarrhea, nausea, vomiting • Connective tissue abnormalities (e.g., joint hypermobility, retained primary teeth)
Testing & Diagnosis	• Serum tryptase levels: determine the level of tryptase in your blood • Genetic testing: determine the number of copies of the tryptase gene (**Alpha-tryptase Copy Number Variation** test offered by Virant Diagnostics)

Systemic Mastocytosis (SM)		
Description & Cause	**Systemic mastocytosis** (SM) is a condition with too many mast cells in multiple parts of the body. The most common cause of SM (~95% of cases) is a mutation that leads to increased numbers of mast cells. The extra mast cells produce extra mediators, such as histamine and tryptase, causing inflammatory responses and SM symptoms.	
Symptoms	• Anaphylaxis • Skin flushing, itching, hives, swelling • Wheezing, shortness of breath • Sinus congestion, pressure • Palpitations, changes in blood pressure, dizziness, fainting • Abdominal pain, bloating, diarrhea, nausea, vomiting	• Anemia, bleeding disorders • Uterine cramping, bleeding • Bone/muscle pain, osteopenia, osteoporosis • Enlarged liver, spleen, lymph nodes • Headache, brain fog, short memory span • Depression, anxiety, mood changes, problem concentrating
Testing & Diagnosis	• Biopsy: examine the mast cells in your skin and/or bone • Serum tryptase levels: determine the level of tryptase in your blood • Genetic testing: detect the SM-causing mutation (**High Sensitivity *KIT* D816V Mutation Hotspot** test offered by Virant Diagnostics)	

If you do not meet the criteria for a diagnosis of HαT or SM, your healthcare provider may consider other MCDs:

- **Cutaneous mastocytosis** (CM) is a condition with too many mast cells in the skin. Patients with CM present with dark, hive-like skin lesions that are tan or brown and are more prone to severe allergic reactions. However, patients with CM are not affected in body parts other than the skin and therefore do not meet the criteria for SM.
- **Mast cell activation syndrome** (MCAS) is a condition with recurring, severe anaphylaxis caused by mast cell mediators; however, patients do not meet the diagnostic criteria for SM, CM, or HαT.

Visit the Resources page on the Virant Diagnostics website to learn more.
https://virantdx.com/resources/