

## Mastocytosis and Other Mast Cell Disorders Frequently Asked Questions

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### Q 1: What are mast cells?

Mast cells are a type of white blood cell located all through the body, but are generally higher in number in the skin, lungs, and intestines. Mast cells form an important part of your immune system. When they are activated (usually in response to a bacteria or virus) they release substances called mediators (histamine, heparin, cytokines and growth factors) causing inflammation. This inflammation protects the body from the harm caused by infection from bacteria and viruses and is also able to reduce the toxicity of some venoms.

Mast cells may also be activated by other triggers such as exercise, temperature changes, some medications and allergy.

When activated during an allergic reaction, the mediators released by mast cells may cause symptoms such as skin itch and flushing, swelling, low blood pressure, fast heart rate, abdominal cramps, diarrhoea, and wheeze. These symptoms may be mild to moderate, however, in severe allergic reaction (anaphylaxis), they can be life threatening.

### Q 2: What are mast cell diseases?

The three major forms of mast cell diseases are:

- Mastocytosis – occurs when the body produces too many mast cells. These cells can continue growing and tend to be overly sensitive to activation and releasing mediators. If the cells build up (accumulate) in organ tissues, this can result in symptoms that affect multiple organ systems.
- Mast Cell Activation Syndrome (MCAS) – occurs when people have signs and symptoms due to mast cells releasing their mediators. Mast cells are normal in number but release an abnormal amount of mediators.
- Hereditary Alpha Trypsinemia (HAT) - an inherited genetic trait where a person has one or more extra copies of the tryptase gene. Tryptase is one of the mediators released by activated mast cells, and people with HAT are known to have higher tryptase levels than normal, even when they are well. People with HAT may have symptoms such as itch, flushing, irritable bowel symptoms or anaphylaxis, but some people with HAT have no symptoms at all.

### Q 3: What is mastocytosis?

Mastocytosis is an abnormal accumulation of mast cells in one or more organ systems. There are three types:

- **Cutaneous mastocytosis (CM)** is considered a benign (mild) skin disease that occurs mostly in children. In 67-80% of cases, resolution will occur before, or in early adulthood.
- **Systemic mastocytosis (SM)** is mostly diagnosed in adults. Mast cells accumulate in the bone marrow and other organs, sometimes including the skin.

- **Mast cell sarcoma (MCS)** is a very rare form of mastocytosis with solid tumour(s) comprising malignant mast cells.

### Q 4: How is mastocytosis diagnosed?

Cutaneous mastocytosis (CM) is diagnosed by the presence of typical skin lesions and a positive skin biopsy with clusters of mast cells.

Systemic mastocytosis (SM) is usually diagnosed by a bone marrow (BM) biopsy. Tryptase levels when measured on a blood test are almost always high in people with SM.

### Q 5: What are mast cell activation syndromes?

People with a mast cell activation syndrome (MCAS) experience repeated episodes of symptoms due to release of mast cell mediators. Symptoms are variable and can include hives, wheeze, gastrointestinal upset, low blood pressure, and anaphylaxis. High levels of mast cell mediators are released during these episodes.

These episodes usually respond to treatment with inhibitors or blockers of mast cell mediators.

### Q 6: How are mast cell activation syndromes diagnosed?

Diagnosis of MCAS usually involves measurement of mast cell mediators, which increase during the episode. For example, a tryptase level measured on a blood test may be high during an episode, and then return to lower levels after the episode.

Other tests that detect a rise in mediator levels have limitations and many are not commercially available in Australia and New Zealand.

A trial of treatment using inhibitors of mast cell mediators can assist diagnosis if symptoms improve during treatment.

### Q 7: How are mastocytosis and mast cell activation syndromes managed?

People who have frequent reactions as a result of mastocytosis or mast cell activation syndromes need ongoing management by a doctor, which should include:

- Referral to a clinical immunology/allergy specialist [www.allergy.org.au/patients/locate-a-specialist](http://www.allergy.org.au/patients/locate-a-specialist)
- Adrenaline injector prescription if required (EpiPen® or Anapen®).
- Regular follow up visits to a clinical immunology/allergy specialist.
- Strategies to avoid triggers if known.

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