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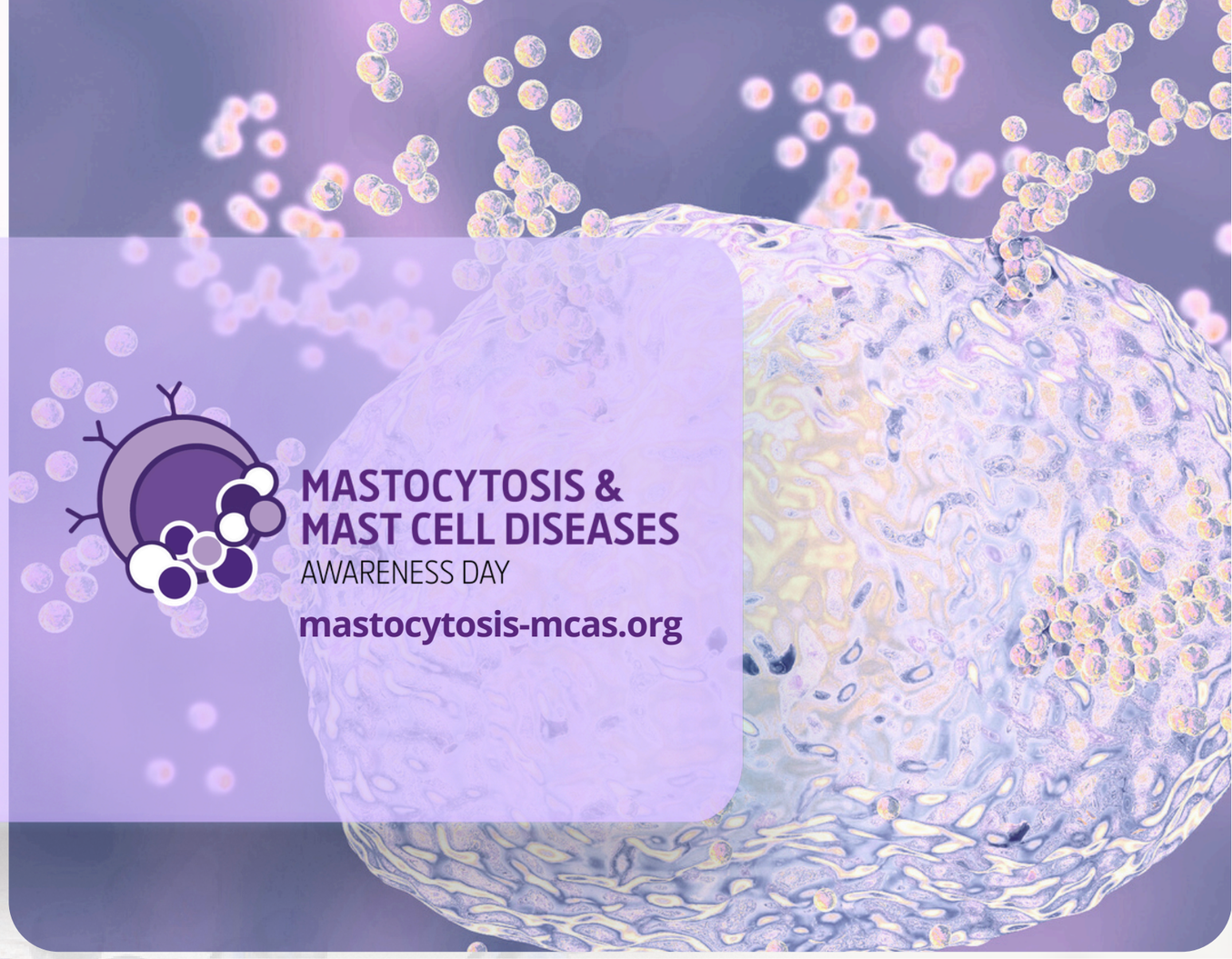
MAST CELL DISEASES

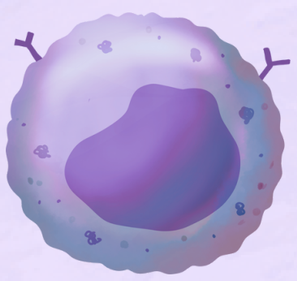
I suspect I have a mast cell disease

A first-step guide for patients, families and communities



MASTOCYTOSIS &
MAST CELL DISEASES
AWARENESS DAY
mastocytosis-mcas.org

What are mast cell diseases?



Mast cell diseases are part of the immune system. They're found in most tissues throughout the body, especially those that interact with the outside world, including the lungs, gastrointestinal tract and the skin.

When a mast cell is activated by a trigger, it releases many mediators (chemicals that set off processes that lead to symptoms). Histamine is one of 200+ mediators which mast cells may release.

Mast cell diseases happen when these cells aren't behaving normally, and reactions can be amplified in people who have too many mast cells.

Types of mast cell diseases:

Mastocytosis is a condition where your body has an unusual buildup of mast cells in one or more parts. Mast cells help protect you, but when you have mastocytosis, these cells misbehave. There are four main groups: one that affects only the skin **(cutaneous mastocytosis)**, one that can affect organs such as the bone marrow and GI tract and other organs **(non-advanced systemic mastocytosis)**, and two that more seriously affect the function of key body systems **(advanced systemic mastocytosis (AdvSM)** and **mast cell sarcoma**).

People with **mast cell activation syndrome (MCAS)** have recurring episodes where their mast cells release mediators, leading to a range of symptoms. These symptoms can be quite diverse, including hives, wheezing, gastrointestinal discomfort, low blood pressure and even severe reactions like anaphylaxis. Fortunately, these episodes often improve with the help of medications that can block or inhibit these mast cell mediators. Diagnosing MCAS isn't always straightforward; it's like solving a medical puzzle for doctors. They consider when the person experiences symptoms, how well medications work, and specialised tests to piece together the full picture and effectively manage this condition.

Hereditary alpha tryptasemia (HαT) is a genetic trait found in about 5 per cent of the studied populations. It's caused by having extra copies of a gene called TPSAB1, which makes a protein called α-tryptase. HαT can lead to various symptoms, although it's tough to pinpoint so far if it directly causes them. Symptomatic people with HαT can experience mild to severely debilitating symptoms, similar to other mast cell diseases. HαT can occur with another mast cell disease and can increase the risk of severe anaphylaxis. Surprisingly, up to two-thirds of people with the HαT trait might not experience noticeable symptoms.

Symptoms can differ between patients. Mast cell diseases can occur in both adults and children.

Common symptoms

Symptoms vary widely and may affect multiple body systems. They may be triggered by stress, illness or environmental exposures.

Possible symptoms include:

- Skin: hives, flushing, itching
- GI: nausea, diarrhea, abdominal pain
- Respiratory: wheezing, nasal congestion
- Cardiovascular: dizziness, low blood pressure
- Neurological: brain fog, headaches
- Systemic: fatigue, temperature intolerance

Common triggers

Each person with a mast cell disease is different – some may react to the following triggers, while others may not react to any.

Mast cells may react to:

- Heat, cold or pressure
- Infections or surgery
- Some foods
- Fragrances and environmental factors
- Medications
- Stress or hormonal changes

What to do next

Track your symptoms:

- Keep a diary noting symptoms, triggers and timing, if necessary
- Record any medications or supplements taken

See a doctor:

- Ask about mast cell diseases.
- Discuss testing for mast cell mediators
- Consider referral to experienced haematologist, immunologist or allergist

Treatment may include:

- H1 and H2 antihistamines
- Mast cell stabilisers
- Leukotriene receptor antagonists
- Lifestyle and dietary adjustments
- In advanced forms of mast cell disease, targeted medicines or stem cell transplants may help lower mast cell burden or replace abnormal cells.

When to seek emergency help

Call emergency services if you experience:

- Difficulty breathing
- Swelling of the face, lips or throat
- Sudden drop in blood pressure
- Severe diarrhea or vomiting
- Loss of consciousness

References

Personalized Management Strategies in Mast Cell Disorders: ECNM-AIM User's Guide for Daily Clinical Practice. The Journal of Allergy and Clinical Immunology: In Practice (2022)

AAAAI Mast Cell Disorders Committee Work Group Report: Mast cell activation syndrome (MCAS) diagnosis and management. J Allergy Clin Immunol (2019)

Hereditary alpha-tryptasemia and monoclonal mast cell disorders. Frontiers in Allergy (2025).

Mast Cell Diseases in Practice and Research: Issues and Perspectives Raised by Patients and Their Recommendations to the Scientific Community and Beyond. The Journal of Allergy and Clinical Immunology: In Practice (2022)

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