AN INTRODUCTION TO SYSTEMIC MASTOCYTOSIS

Types, symptoms, diagnosis, and questions to ask your doctor

What is systemic mastocytosis?

Systemic mastocytosis (pronounced mass-toe-sigh-TOE-sis), or SM for short, is a rare disorder that causes the body to make too many abnormal mast cells. Mast cells are white blood cells that aid the immune system and help fight off illness. When these cells are activated, it can result in symptoms similar to an allergic reaction. A severe allergic reaction, known as anaphylaxis, can cause severe inflammation and organ damage.

THERE ARE 2 MAIN TYPES: ADVANCED SYSTEMIC MASTOCYTOSIS AND NONADVANCED SYSTEMIC MASTOCYTOSIS

ADVANCED SYSTEMIC MASTOCYTOSIS

A life-threatening disease that may be associated with a shortened life expectancy due to infiltration of mast cells into major organs

Symptoms may include:

- Fatigue
- Nausea, malabsorption, and weight loss
- Low blood counts
- Bone pain and/or fractures
- Abnormal liver function
- Liver and/or spleen enlargement

NONADVANCED SYSTEMIC MASTOCYTOSIS

A lifelong disease associated with poor quality of life due to debilitating symptoms and sometimes life-threatening allergic reactions

Symptoms may include:

- Itching, skin lesions, skin redness, or warmth (flushing)
- Wheezing, shortness of breath
- Fatigue
- Allergic reaction
- Abdominal pain, bloating, diarrhea, nausea, vomiting
- Brain fog, dizziness, memory issues, mood changes, or trouble concentrating
- Bone pain and/or fractures

SYSTEMIC MASTOCYTOSIS is different from other mast cell disorders, such as mast cell activation syndrome (MCAS) or cutaneous mastocytosis (CM), because it occurs when the body produces too many abnormal mast cells that accumulate in the bone marrow and different organs. An **allergist/immunologist** and/or **hematologist** may be able to help diagnose mastocytosis.

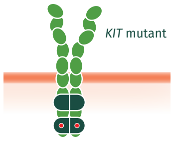
DIAGNOSING SYSTEMIC MASTOCYTOSIS

Systemic mastocytosis is a rare disease that shares symptoms with other, more common disorders. This can make it difficult to diagnose systemic mastocytosis and may lead to misdiagnoses.

THERE ARE TESTS THAT CAN HELP DIAGNOSE SYSTEMIC MASTOCYTOSIS

Tryptase test

- **What it does:** measures the amount of tryptase in your blood (tryptase is an enzyme released by mast cells and is increased in systemic mastocytosis)
- **How it's performed:** a doctor or lab worker will take a sample of your blood
- **Where it's done:** a doctor's office, lab facility, or emergency room
- **How long will it take for my results?** About 1 to 2 days



KIT D816V blood test

- **What it does:** identifies a genetic mutation that has been found in about 95% of people with systemic mastocytosis
- **How it's performed:** a doctor will order a high-sensitivity KIT D816V blood test and draw your blood
- **Where it's done:** a doctor's office or a lab facility
- **How long will it take for my results?** About 5 to 7 days

Biopsy

- **What it does:** helps to confirm the diagnosis of systemic mastocytosis by identifying abnormal mast cells in the tissue
- **How it's performed:** a doctor or lab worker will take a small sample of your skin, bone marrow, or other organ involved with systemic mastocytosis
- **Where it's done:** a doctor's office or a lab facility
- **How long will it take for my results?** Varies based on the type of biopsy and center; always discuss timelines for results with your doctor

ONLY A DOCTOR CAN DIAGNOSE SYSTEMIC MASTOCYTOSIS AND DETERMINE WHICH TESTS ARE APPROPRIATE

If you think you might have systemic mastocytosis, make an appointment with your doctor today. **Remember to bring this resource with you to the appointment.**

TALK TO YOUR DOCTOR ABOUT SYSTEMIC MASTOCYTOSIS

Be sure to share the following information:

- Your symptoms such as unexplained fatigue, brain fog, shortness of breath, bone or muscle pain, abdominal pain, diarrhea, nausea, allergic reactions, anaphylaxis, and other symptoms you may be experiencing. **It can help to keep a journal and bring it with you to your appointment**
- The symptoms of your disease can be triggered by emotional, physical, or environmental factors that may affect a change in your symptoms, or allergic reactions
- Your medical history (including any conditions or symptoms unrelated to systemic mastocytosis)

BE SURE TO ASK YOUR DOCTOR ABOUT TESTING AND NEXT STEPS. HERE ARE SOME QUESTIONS TO HELP START THE CONVERSATION:

- **Should I see any other specialists (allergist/immunologist or hematologist) about my symptoms?**
- **How can I schedule any follow-up test(s) or evaluations?**
- **Can you order a serum tryptase test for me?**
- **How do I access a KIT D816V blood test?**
- **Should we schedule a biopsy to look for mast cell accumulation?**
- **How can I manage my symptoms while I wait for test results?**
- **If systemic mastocytosis isn't the cause of my symptoms, how can we determine what is?**
- **Can you recommend any resources for more information, or support groups for people living with systemic mastocytosis?**

As systemic mastocytosis is rare, many doctors or specialists may not be familiar with the disease. It's important to advocate for yourself and ask your doctor if you should be evaluated for systemic mastocytosis. It could help you get the answers you need for an accurate diagnosis and appropriate medical care.

If you've been diagnosed with systemic mastocytosis and are interested in participating in one of our clinical trials, visit our website to learn more.

www.mastocytosistrials.com/DX

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BIOSCIENCES

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