Diagnosing Systemic Mastocytosis?

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International Foundation for Gastrointestinal Disorders (www.iffgd.org)

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What Is Systemic Mastocytosis?

Systemic Mastocytosis is a rare group of conditions where the body makes too many mast cells. Mast cells are a type of immune cell that help protect us from infections by releasing chemicals like histamine to generate an immune response. However, when there are too many mast cells, they can release these chemicals in large amounts. This usually results in symptoms like those found in allergic reactions. Some of these may be potentially life-threatening and known as anaphylactic reactions. Symptoms of anaphylaxis include swelling, hives, difficulty breathing, and feeling of passing out.

Hives – welts on the skin that are raised and itch.

How do we diagnose Systemic Mastocytosis?

Your provider may suspect this diagnosis when you first discuss symptoms, and they have examined you. Typical symptoms can range from skin (hives, flushing, or itching) to gastrointestinal (intermittent abdominal pain, nausea, or diarrhea). There are also other symptoms suggesting Systemic Mastocytosis such as fatigue, bone pain, palpitations, and throat closing sensations with reactions.

Your provider will next order blood tests that are intended to look for signs of the disease but also potential early complications. When trying to initially diagnose Systemic Mastocytosis, labs should ONLY be obtained when you are at baseline health, not after a flare of symptoms, and can include:

- **Tryptase level:** Tryptase is an enzyme released by mast cells. High levels of tryptase (greater than 20 ng/mL) are a major sign that systemic mastocytosis may be the cause of the symptoms.
- **Complete blood count:** to look for abnormalities in blood counts, like red and white blood cells.
- **Liver tests:** to look for involvement of Mastocytosis in your liver where abnormal mast cells can accumulate. This may also be called a liver function test.
- **Genetic testing of the KIT Gene:** KIT is a receptor found on the surface of mast cells and is responsible for the production of mast cells. Systemic Mastocytosis most commonly occurs when there is a mutation in KIT; a specific blood test is able to test for this mutation which can be a key part of the diagnosis.

Learn more about the symptoms of Systemic Mastocytosis with IFFGD Fact Sheet No. 903

If any of these tests are abnormal, then additional testing will be needed to definitively diagnose Systemic Mastocytosis. Based on guidance from the World Health Organization (WHO), tissue sampling, also known as biopsies, are required to confirm the diagnosis. Biopsies can be obtained from the following areas for testing:

- Bone marrow (preferred option)
- Skin
- Gastrointestinal tract - This can be done via upper endoscopy– an examination of the upper GI tract including the stomach and first part of the small intestine or colonoscopy to assess the colon.

If the biopsy shows there is an abnormal number of mast cells – specifically, at least 15 cells that are clustered together – then this is sufficient to make the diagnosis. However, mast cell clusters may not be seen under the microscope in some patients, possibly because the tissue sample only gives us a small picture as to what is going on. In these situations, three of the following findings may still permit a diagnosis of Systemic Mastocytosis:

- Biopsy (from any tissue source listed above) that shows that at least 25% of the mast cells are considered abnormal in appearance.
- DNA testing that finds a specific mutation in the KIT gene (which is important for the production of mast cells).
- The mast cells in the biopsy express a specific marker on the cell surface called CD25, which tells us that there is an abnormal population of mast cells in the tissue that was biopsied.
- The blood test for "tryptase" is elevated above the normal level (greater than 20 ng/mL)

If you are ultimately diagnosed with Systemic Mastocytosis, then there are additional tests that may be needed to determine which specific type of Mastocytosis you have.

Learn more about the types of Systemic Mastocytosis with IFFGD Fact Sheet No. 904

The World Health Organization (WHO) is a specialized agency of the United Nations focused on international public health. Founded in 1948, its mission is to promote and protect global health by setting health standards, providing leadership, coordinating disease control efforts, and supporting countries in strengthening their health systems. WHO works to ensure health equity, improve access to care, and respond to global health emergencies like pandemics, while promoting research and evidence-based policies to improve health outcomes worldwide.

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The International Foundation for Gastrointestinal Disorders (IFFGD) is a 501(c)(3) nonprofit education and research organization. We work to promote awareness, scientific advancement, and improved care for people affected by chronic digestive conditions. Our mission is to inform, assist, and support people affected by gastrointestinal disorders. Founded in 1991, we rely on donors to carry out our mission. Visit our website at: www.iffgd.org.

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