

No Disease Too Small - Patient Guide

*No Disease Too **S**mall* is a campaign that offers information and guidance for patients and their loved ones as they search for answers about symptoms and the disease. An educated patient and physician can work together as partners throughout the diagnosis and treatment journey.

Download the patient guide (PDF 1.0 MB)

Accordion:

A guide to systemic mastocytosis (SM)

Systemic mastocytosis (SM) is a group of rare blood diseases where too many abnormal mast cells, which are specialized immune cells, collect in more than one body tissue. Mast cells can accumulate in the skin and internal organs, including the liver, spleen, bone marrow, and tissues of the gastrointestinal tract, which can lead to inflammatory and allergic reactions and other symptoms^{1,2,3}.

What is the clinical definition of mastocytosis?

- Mastocytosis is a rare disease where the body makes abnormal mast cells that accumulate in various organ systems^{2,4}.
- A “mast” cell is a type of white blood cell in the immune system².
 - Mast cells help heal wounds and protect the body from infection. Mast cells also are specialized to release multiple chemical mediators, including histamine, tryptase, heparin and others, which can contribute to inflammatory and allergic reactions and other clinical symptoms^{5,6}.
- “Cytosis” is a suffix that means an increase in the number of cells⁷.
- There are two types of mastocytosis: systemic and cutaneous^{2*}.
- In SM, the accumulation of mast cells can affect both the skin and internal organs, resulting in systemic symptoms, and may cause organ damage^{1,3}.

**Cutaneous mastocytosis is more common in children, and is outside the scope of this document².*

How common is SM?

- SM is very rare, and people who have SM may have difficulty finding support^{1,4}.
- Around the world, SM affects between 1 in 20,000 and 1 in 40,000 people¹.
- The median age at SM diagnosis in adults is 55 years⁸.
- SM is not usually linked to family history; the risk of getting SM is greater if there is a

change in a particular gene³.

- Blood tests that measure levels of tryptase, as well as bone marrow tests, are necessary to diagnose SM³.

Why is SM so difficult to diagnose?

- Symptoms may initially be minor and progress over time.
- SM is underdiagnosed for several reasons:
 - Symptoms frequently seen in SM are often the same as those seen in many other illnesses, frequently leading to incorrect diagnosis⁸.
 - Physicians are not accustomed to seeing the disease. Patients may cycle between many different physicians and specialists before receiving a correct diagnosis⁴.
- An SM diagnosis can take 2 to 10 years⁴.

What impact does SM have on the body?

- There are different kinds of SM, and some are more serious than others. Very serious cases can lead to organ damage or even organ failure³.
- When the body produces too many mast cells, the increased number of chemical mediators released can result in symptoms, including, but not limited to, flushing, itching, nausea, diarrhea, low blood pressure and anaphylaxis^{2,3,6}.
- The organs SM affects are usually the bone marrow, skin, liver, spleen, lymph nodes, and those of the gastrointestinal tract³.
- Symptoms may depend on which organ is being affected:
 - Bone involvement can result in bone pain⁶.
 - Nausea, vomiting, abdominal pain, and diarrhea are seen when cells in the gastrointestinal (GI) tract are affected⁹.
 - When mast cells begin to accumulate in body tissues, the lymph nodes, liver, and spleen may swell^{3,6}.
 - When the skin is affected, symptoms including severe itching and flushing (which can be triggered by temperature changes), fever, exercise and friction⁶.
- Physicians should work to develop a treatment plan best suited to manage patient symptoms¹⁰.

What are the symptoms of SM?

- SM symptoms can vary significantly from one person to another.
- If the following symptoms are present, the patient should talk to a health care provider^{2,3,9,11}.
 - Skin rash
 - Itching or pruritus
 - Flushing of the skin
 - Anemia
 - Digestive problems such as stomach pain, diarrhea, malabsorption, weight loss, nausea, or vomiting
 - Extreme allergic-like reactions called anaphylaxis that can occur after a bee or insect sting that can cause swelling of the face, tongue, or eyes; trouble breathing, or dizziness

- Swelling in liver, spleen, or lymph nodes
- Headache
- Fatigue
- Osteoporosis
- Bone pain
- Extreme reactions to heat, cold, or other physical triggers

Misdiagnoses of other diseases

- People with SM are often diagnosed with other, more common illnesses because they have the same symptoms as SM, including⁸.
 - Irritable bowel syndrome
 - Malabsorption syndrome
 - Urticaria
 - Carcinoid syndrome
 - VIPoma
 - Zollinger-Ellison syndrome
 - Leukemia
 - Other myeloproliferative disorders

If a patient has been diagnosed with chronic myelomonocytic leukemia (CMML), myelodysplastic syndrome (MDS), myeloproliferative neoplasms (MPN), acute myeloid leukemia (AML), myeloproliferative disorder (MPD), non-Hodgkin's lymphoma (NHL) or hypereosinophilic syndrome (HES) and the above symptoms are present, the individual could also have a mast cell disorder²⁴.

- When a diagnosis of SM is made, a patient may have visited other types of health care providers to find relief for their symptoms, including:
 - Dermatologists
 - Allergists
 - Hematologists
 - Gastroenterologists
 - Endocrinologists
 - Internal medicine specialists
 - Rheumatologists
 - Cardiologists
 - Neurologists

How does SM impact someone's everyday life?

Like many other long-term or chronic diseases, SM can dramatically affect a person's daily quality of life¹¹.

- Living with a chronic illness, especially one so difficult to diagnose, can impact the mental health of the patient¹¹.
- Some of the feelings a patient with SM may feel during their diagnosis and treatment are

:

- Increased stress
- Fear and/or anxiety
- Poor motivation
- Sadness and/or depression
- Isolation
- If a patient with SM is experiencing physical or emotional health problems, he or she should talk with a health care professional. It is important to seek support and counseling to help cope with these feelings.

If a patient is concerned about SM, what should he or she do?

- An informed conversation with a physician is the first step. Patients with symptoms common in SM should not hesitate to ask questions about the disease.
- It is important to know about the different types of SM¹⁰ – there are five – and to ask specific questions about testing that can help a physician make an SM diagnosis, so treatment can begin.
- For a list of key questions to help both patients and health care providers, visit *No Disease Too Small* (www.nodiseasetoosmall.com ^[1]).

The five types of SM

SM is categorized into five subtypes, determined by the affected organs¹⁰. They differ in severity, treatment approach, and life expectancy^{3,10,12}.

- **Indolent SM (ISM)** is the most common and mild type of SM³. Symptoms may include itching skin, fainting, headaches, vomiting, or diarrhea¹³. Those with ISM have a low risk of progression to a more advanced form⁶.
 - Prognosis: Typically favorable⁶.
- **Smoldering SM (SSM)** is usually characterized by a high volume of mast cells in the bone marrow. Those with SSM may have a higher likelihood of progressing to an advanced disease category⁶.
 - Prognosis: May indicate tendency to progress to a more advanced form⁶.
- **SM with an associated hematologic neoplasm (SM-AHN)** is a type of SM where the patient has an additional blood disorder, most often myeloproliferative disorders or a myelodysplastic syndrome⁶.
 - Prognosis: Depends on associated hematological neoplasm⁶.
- **Aggressive SM (ASM)** is a severe type of SM. Mast cells will collect in different organs and can cause organ damage or failure³.
 - Prognosis: Less favorable⁶.
- **Mast cell leukemia (MCL)** is a rare and serious type of SM. There are many more mast cells circulated in the bone marrow with MCL. The outcome with MCL is very poor⁶.
 - Prognosis: Life expectancy may be limited⁶.

SM-AHN, ASM, and MCL are all defined as advanced SM and have lower survival rates compared to ISM and SSM¹⁴.

Which tests can diagnose SM?

- Because SM symptoms are commonly found in other diseases (such as a skin rash), it can be difficult to diagnose⁸.
- There are, however, several specific tests that can more precisely determine if SM is present:
 - **Blood tests** frequently used to diagnose SM:
 - **Blood serum tryptase test:** Serum tryptase is an enzyme, or chemical, produced by the mast cells⁵. A person with SM is likely to have higher levels of tryptase in the bloodstream³.
 - **Other blood tests to evaluate organ damage:**
 - **Blood serum albumin test:** A serum albumin test is a simple blood test that measures the amount of albumin in the patient's blood. Low levels of albumin are often associated with inflammation in the liver, kidney disease and malnutrition¹⁵.
 - **Alkaline phosphatase (ALP) test:** The test measures the amount of ALP enzyme in the bloodstream. If ALP levels are elevated, it indicates a problem with the liver or bones¹⁶.
 - **Hemoglobin test:** Anemia, a condition in which the blood has fewer red blood cells than normal, can be a sign of SM^{3,17}. A hemoglobin test is used to check for anemia¹⁸.
 - **Complete Blood Count (CBC):** Low blood platelet count (thrombocytopenia), and high white blood cell count (leukocytosis) can be signs of advanced variants of SM^{3,20}. CBC tests measure many different parts of the blood, including the overall number of white blood cells and platelets¹⁸.
 - **Genetic mutation testing:** Learning if a genetic mutation is present can help a physician diagnose SM³. A mutation is a change in a gene's structure⁶. In SM, changes in a gene called KIT make mast cells keep growing²⁰. About 9 out of 10 patients with SM have a KIT mutation called KIT D816V²¹. Detecting the genetic mutation is an important part of identifying SM³.
 - **CT Scanning and other imaging:** Infiltration of abdominal organs and lymph nodes can be seen best by a variety of CT and other scans²².
 - **Biopsies**
 - **Bone marrow biopsy:** Mast cells form in the bone marrow⁵. Physicians can test a patient's bone marrow to see if it contains abnormal mast cells, meeting specific criteria. In addition to determining the presence of SM, the test can help physicians understand which type of SM is present³. A bone marrow biopsy is a procedure performed using a biopsy needle to extract organ samples²³.

For more information and resources, visit *No Disease Too **Small*** (www.nodiseasetoosmall.com ^[1]).

The Mastocytosis Society* may be another source of information for you: www.tmsforacure.org

^[2]

Footnotes:

*The Mastocytosis Society is a not-for-profit group, independent from Novartis. Novartis has no financial interest in this organization, but may provide occasional funding support. Novartis is not responsible for the organization's information content or actions.

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Source URL: <https://www.novartis.com/our-focus/cancer/supporting-people-affected-systemic-mastocytosis/no-disease-too-small-patient-guide>

Links

[1] <http://www.nodiseasetoosmall.com>

[2] <https://www.tmsforacure.org/>