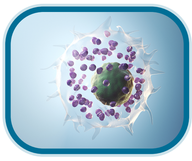
Learning About Systemic Mastocytosis (SM)

What is a mast cell?

Before you learn about SM, it's important to first know what a mast cell is.

A mast cell is a type of white blood cell that is a key part of the immune system. Mast cells are designed to help keep the body safe from diseases and infections.



What causes SM?

If mast cells are abnormal and uncontrolled, they often increase or spread throughout the body, releasing proteins. These proteins may often trigger symptoms of allergic reactions, inflammation, and other immune responses. SM is a rare disease that results from the spreading of these abnormal mast cells.

In about 95% of cases, SM forms because of a genomic change known as *KIT D816V*.[1–3] This change (or mutation) leads to the production of too many abnormal mast cells.[4]

What can happen as a result of SM?

People with SM may experience different symptoms that may be unexplained, unpredictable, recurring, severe, or little-to-no symptoms. The following are some of the potential symptoms of SM:

- Skin rashes, hives, anaphylaxis
- Abdominal pain, diarrhea, nausea
- Shortness of breath, wheezing
- Bone pain, bone health issues
- Unexplained fatigue, weakness
- Low blood pressure, rapid heart rate
- Brain fog, anxiety, and depression

What are the types of SM?

Non-advanced SM
About 85% of SM cases[3,5,6]

Advanced SM
About 15% of SM cases[3,5,6]

Non-advanced SM[7,8]

Non-advanced SM may cause many difficult symptoms. Subtypes of non-advanced SM:

Bone marrow mastocytosis (BMM)
In BMM, abnormal mast cells build up only in the bone marrow, not the skin.

Indolent SM (ISM)
ISM is the most common type of SM. Abnormal mast cells are mainly found in the bone marrow but also in other parts of the body, like the skin.

Smoldering SM (SSM)
In SSM, abnormal mast cells build up in the bone marrow and organs, potentially causing more significant symptoms.

Advanced SM[7,8]

Advanced SM occurs when abnormal mast cells continue to build and form throughout the body. This can lead to potentially life-threatening organ damage. Subtypes of advanced SM:

Aggressive SM (ASM)
A severe form of SM in which abnormal mast cells can commonly be found in the bone, liver, and digestive system.

SM with an associated hematological neoplasm (SM-AHN)
SM-AHN occurs in combination with other blood or bone marrow disorders.

Mast cell leukemia (MCL)
MCL affects the bone marrow and white blood cells. It is extremely rare and can be more serious.

How many people have SM?

SM is a rare disease that affects up to

1 in 5,000 adults[9–12*]

20–50 years old
People with SM may first start to notice signs or symptoms at 20–50 years of age[2]

~6 years
People with SM report averaging 6 years from first noticing symptoms to getting a diagnosis[13]

To gain a better understanding of SM, visit: **www.MastoConnect.com**

*A new study shows that SM might be more common than we previously thought.

References: **1.** García-Montero AC et al. *Blood.* 2006;108(7):2366–2372. **2.** Kristensen T et al. *Am J Hematol.* 2014;89(5):493–498. **3.** Ungerstedt J et al. *Cancers.* 2022;14(16):3942. **4.** Kristensen T, et al. *J Mol Diagn.* 2011;13(2):180–188. **5.** Cohen SS et al. *Br J Haematol.* 2014;166:521–528. **6.** Sperr WR et al. *Lancet Haematol.* 2019;6:e638–e649. **7.** WHO Classification of Tumours Editorial Board. Haematolymphoid tumours [Internet]. Lyon (France): International Agency for Research on Cancer; 2024 [cited April 24, 2024]. (*WHO Classification of Tumours Series*, 5th ed.; vol. 11). Available from: https://tumourclassification.iarc.who.int/chapters/63 **8.** Arber DA et al. *Blood.* 2022;140(11):1200–1228. **9.** Bergström A, et al. *Acta Oncol.* 2024;63:44–50. **10.** Cohen SS, et al. *Br J Haematol.* 2014;166: 521–528. **11.** Van Doormaal JJ, et al. *J Allergy Clin Immunol.* 2013;131:1429–31. **12.** Brockow K. *Immunol Allergy Clin North Am.* 2014;34(20):283–295. **13.** Mesa RA et al. *Cancer.* 2022;128(20):3691–3699.

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