Types of Systemic Mastocytosis?

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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.

Types of Systemic Mastocytosis

There are several types of Systemic Mastocytosis. They differ in how many mast cells build up, where they build up in the various parts of the body, and the types of symptoms that may be experienced. Importantly all must satisfy the specific diagnostic criteria for Systemic Mastocytosis:

- Indolent Systemic Mastocytosis (ISM)
- Smoldering Systemic Mastocytosis (SSM)
- Systemic Mastocytosis with an Associated Hematologic Neoplasm (SM-AHN)
- Aggressive Systemic Mastocytosis (ASM)
- Mast Cell Leukemia (MCL)

Indolent Systemic Mastocytosis (ISM)

This form of Systemic Mastocytosis is the most common and has a milder course. With fewer than 20% of mast cells in the bone marrow, this form does not impair how the organs function. People with ISM will still experience symptoms of mast cell activation and are at risk for complications including anaphylaxis and bone disease (osteoporosis).

Smoldering Systemic Mastocytosis (SSM)

This is a higher risk subtype of Systemic Mastocytosis with a chance of progressing to a more advanced form. Similar to the ISM subtype, the SSM subtype will have fewer than 20% of mast cells in bone marrow. While the organs all continue to work properly in SSM, there are large infiltrations of the abnormal mast cells in organs leading to enlargement in some cases and very high tryptase levels.

Systemic Mastocytosis with an Associated Hematologic Neoplasm (SM-AHN)

This form of Systemic Mastocytosis is the only form where there is another blood cancer present at the same time. The SM-AHN subtype is similar to the previous subtypes mentioned in that there are fewer than 20% of mast cells in the bone marrow without organ dysfunction. However, the presence of another blood cancer may significantly impact the disease course and mortality risk. Common associated blood cancers include acute myeloid leukemia and myelodysplastic syndrome where the bone marrow doesn't function to make the blood cells.

Aggressive Systemic Mastocytosis (ASM)

Like the name suggests, this form of Systemic Mastocytosis is more severe. Without a blood cancer and fewer than 20% of mast cells in bone marrow, this form may appear like the others. However, patients with ASM have organ dysfunction due to the mass infiltration of abnormal mast cells. They may experience problems with intestinal absorption, and liver and bone marrow dysfunction.

Mast Cell Leukemia (MCL)

This is the rarest and most aggressive form of Systemic Mastocytosis that typically requires immediate treatment. It may progress quickly and has a right risk of death. Unlike the other forms of Systemic Mastocystosis Mast Cell Leukemia will have 20% of mast cells or more in bone marrow.

Identifying Subtypes

To identify the potential subtypes there are certain tests and clinical criteria that must be met. Of the following criteria listed below, at least one will be seen with the Aggressive Systemic Mastocytosis (ASM) subtype and **none** will be seen with the Indolent Systemic Mastocytosis (ISM) subtype:

- Abnormally low blood cell counts but no signs of a blood cancer.
- Enlarged liver with signs of abnormal liver function (e.g. abnormal labs, abdominal fluid)
- Abnormal bone scans suggesting significant bone loss that may increase risk for bone fractures
- Evidence of poor absorption of nutrients and fluids through the gastrointestinal tract with weight loss, diarrhea and evidence of malnutrition

For the Smoldering Systemic Mastocytosis (SSM) subtype none of the previously listed criteria will be seen but **two** of the listed criteria will be present:

- Bone marrow biopsy shows more than 30% mast cells (mast cells normally compose <1% of the cells in the bone marrow) and tryptase level over 200 ng/mL
- Signs of abnormal looking blood cell types but blood cell counts are normal or only slightly abnormal
- Enlarged liver, spleen, or lymph nodes (but with normal organ functioning on lab tests)

For the Mast Cell Leukemia (MCL) subtype, a bone marrow biopsy will show atypical and immature forms of mast cells that comprise at least 20% of the total cells. Findings on testing that suggest a secondary blood cell disorder or cancer along with Systemic Mastocytosis would suggest Systemic Mastocytosis with an Associated Hematologic Neoplasm (SM-AHN) subtype.

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