Understanding advanced systemic mastocytosis (SM)

Advanced SM is a group of rare diseases in which uncontrolled growth and accumulation of mast cells (a type of white blood cell) occurs in one or more organs, including the GI tract, spleen, lymph nodes and bone marrow.\(^1\)

Mast cells release substances, such as histamine, which can lead to symptoms including itching, fever, abdominal pain, nausea and vomiting.\(^2\) The uncontrolled growth and reproduction of mast cells is caused in many people by a KIT mutation.\(^3\) KIT D816V is the most common mutation in SM, occurring in about 90% of patients.\(^4\)

In advanced SM, mast cells collect in such high quantities that they lead to organ damage and dysfunction, bone fractures and anemia.\(^5\)

Advanced SM subtypes are associated with decreased life expectancy compared to people without the disease.\(^6\) The subtypes of advanced SM include:

- **Aggressive systemic mastocytosis (ASM):** Characterized by findings such as low blood cell count, enlarged spleen, decreased bone density leading to fractures, etc.\(^7\) It is caused by growth and accumulation of mast cells in diverse organs, leading to impaired organ function.\(^2\) Median overall survival is 3.5 years.\(^6\)
- **SM with an associated hematologic neoplasm (SM-AHM):** In addition to SM, some patients have an additional blood disorder such as myelodysplastic syndromes or myeloproliferative disorders.\(^2\) In these cases, median overall survival is 2 years.\(^6\)
- **Mast cell leukemia (MCL):** Characterized by findings such as infiltration of the abnormal mast cells in bone marrow, blood, and other organs, with bone marrow aspirate showing mast cells accounting for 20% or more out of all nucleated cells in the bone marrow.\(^1\) May occur de novo or secondary to previous mastocytosis.\(^8\) It is characterized by the rapid accumulation of mast cells in the bone marrow, ultimately resulting in multi-organ failure.\(^9\) Median overall survival is less than 6 months.\(^6\)

Due to their rarity, the exact prevalence of these advanced subtypes of SM is unknown. Treatment is tailored towards each individual patient and their disease.\(^10\) The major goal of treatment is to control mast cell growth and expansion.\(^10\) Another important part of treatment is controlling a person's symptoms.\(^2\)

Questions to ask your doctor

- What type of mastocytosis do I have?
• What is my prognosis and how will advanced SM impact my quality of life?
• Is mastocytosis a type of cancer?
• What are my treatment options?
• What are the risks associated with each treatment option?
• What are the side effects of each treatment?
• Will my symptoms go away after I start treatment?
• How will I know if my treatment is working?
• If my disease stops responding to a course of treatment, what are my options?
• Should I consider clinical trials?
• Are there support groups I can join?

Additional resources

• Systemic Mastocytosis Fact Sheet (PDF 0.5 MB) [2]

Advanced systemic mastocytosis (SM) is a group of rare and fatal diseases of the organs and bone marrow. Learn more about the various subtypes of advanced SM.

Footnotes:

References


Source URL: https://www.novartis.com/our-focus/cancer/oncology-disease-areas/advanced-systemic-
mastocytosis

Links