

Delayed Diagnosis

“Patients often have to move doctor to doctor to find answers.”

- Symptoms are complex and vary person to person, making SM difficult to recognize
- Getting to a specialist is difficult
 - Lack of disease awareness and symptom recognition among general practitioners (GPs)
 - Few specialists and lack of referrals
- Inconsistency of availability and frequency of *KIT* mutation testing among healthcare providers (HCPs)



“When diagnosis is received quickly, like at a reference center, it’s easier to handle symptoms, otherwise, patients have to do a pilgrimage from one doctor to another.”

Lifestyle Burden and Impact on Productivity



“Unpredictability is really difficult for family and friends and work.”

- Unpredictable symptoms disrupt daily activities
- Significant lifestyle adjustments to avoid triggers and manage SM
- Current approach to treatment involves many medications
- Strained social and familial relationships
 - Lack of understanding
 - Chronic nature of illness
 - Need for ongoing support
- Expectations to perform “normally” in the workplace

Lack of Awareness

- Family, friends, employers, and the general public do not understand SM and its impact
- Few healthcare professionals are trained to recognize the signs and symptoms of the disease

“The diagnostic journey is quite long because doctors think we are ‘crazy’ at the beginning.”



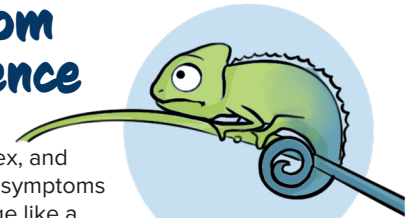
CHALLENGES FOR PEOPLE LIVING WITH SYSTEMIC MASTOCYTOSIS (SM)



“It’s really hard in work where you’ve worked the same place a long time. It’s hard to see that one day you may be really productive and one day you may really be unproductive.”

Symptom Experience

- Varied, complex, and unpredictable symptoms that can change like a chameleon
- Severity of symptoms misunderstood
- No standard of care
- “Invisible” symptoms



“On one day, I may have problems with my stomach, but the next day it’s fatigue, or the following day it’s brain fog—symptoms change day by day and make it very difficult to understand the condition.”

Psychological Impact

“I can be ok in one moment, but in the next moment I could need help because I’m in an emergency situation.”

- Chronic nature of the disease and difficulty controlling symptoms leads to distress, anxiety, and depression
- Patients often feel invisible, crazy, frustrated, and helpless
- Social isolation is common for people with SM
- Frustration at lack of diagnosis

“I think the problem is that it’s not a visible condition. Very often people look very good. For example, my phenomenon is that I either look terrible or I don’t look terrible at all.”

