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 recognise
- 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."

- Significant lifestyle adjustments to avoid triggers and manage SM
- many medications

- Chronic nature of illness

public do not understand SM and its impact Few healthcare professionals are trained to recognize the signs and symptoms of the disease

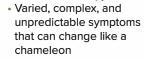
Family, friends, employers, and the general

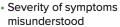
Lack of Awareness

"The diagnostic journey is quite long because doctors think we are 'crazy' at the beginning."









- · 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

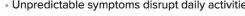
"I can be ok in one moment, but in the next moment I could need help because I'm in an emergency situation."

- 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. Verv often people look very good. For example. mv phenomenon is that I either look terrible or I don't look terrible at all."







- Current approach to treatment involves
- Strained social and familial relationships
- Lack of understanding
- Need for ongoing support
- Expectations to perform "normally" in the workplace

"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."



This material has been developed and funded by Blueprint Medicines. The content summarizes the key insights gained during a virtual International Advisory Board hosted in November 2023. It is important to note that these insights represent opinions and should not be interpreted as educational or medical advice. Participants included representatives from the following patient organizations: Asociación Española de Mastocitosis y Enfermedades Relacionadas (AEDM), Associação Portuguesa de Doentes com Mastocitose e Doenças Mastocitárias (APORMast), Association française des patients atteints de mastocytoses et de syndromes d'activation mastocytaires (ASSOMAST), Selbsthilfeverein Mastocytoses et V., Associazione Italiana Mastocitosi (ASIMAS), Mastozytose Selbsthilfe Netzwerk e.V, SELMAS mastozytose.ch, and The UK Mastocytosis Support Group.

