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."

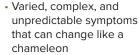
- Unpredictable symptoms disrupt daily activities
- Significant lifestyle adjustments to avoid triggers and manage SM
- Current approach to treatment involves many medications
- Strained social and familial relationships
- Lack of understanding
- Chronic nature of illness
- Need for ongoing support
-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

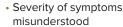
"It's really hard in work where you've worked the same place a long time. It's hard to see that one day you

may be really productive and one day you may really be unproductive."

"I keep explaining, explaining, explaining, and often feel helpless."







- · No standard of care
- "Invisible" symptoms





"On one day, I may have problems with my stomach, but the next day it's fatique, 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, crazv, 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."



Lack of Awareness

· Family, friends, employers, and the general

Few healthcare professionals are trained to

public do not understand SM and its impact

recognize the signs and symptoms of the disease

"The diagnostic journey is quite

long because doctors think we

are 'crazy' at the beginning."



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 (AEDMI), Associação Portuguesa de Doentes com Mastocitose e Doencas Mastocitárias (APORMast), Association française des patients atteints de mastocytoses et de syndromes d'activation mastocytaires (ASSOMAS), Association Mastozytose e.V., Associazione Italiana Mastocitosi ONLUS (ASIMAS), Mastozytose Selbsthilfe Netzwerk e.V, SELbsthilfenetz MAstozytose Schweiz, and The UK Mastocytosis Support Group.

