When One Is Diagnosed, Two Are Changed: The Caregiver Experience in Indolent Systemic Mastocytosis

Poster Number 513

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Introduction

- Indolent systemic mastocytosis (ISM) is a clonal mast cell disease driven by the *KIT* D816V mutation in ~95% of adult cases[1–3]
- People living with ISM can experience lifelong, life-altering symptoms across multiple organ systems, often shaping every part of their daily lives[4–8]
- Most individuals rely on polypharmacy for the management of symptoms with best supportive care (BSC) medications, although in many individuals, symptoms are not adequately controlled with BSC medications[8–9]
- In ISM, the burden extends far beyond the person living with ISM, altering daily life for caregivers who function as advocates, coordinators, and safety monitors within complex and often fragmented care systems
- There is meaningful opportunity to better integrate caregivers' perspectives into care models to include symptom monitoring, care coordination, and crisis preparedness
- Caregivers were engaged to gather in-depth qualitative insights into the current situation for caregivers in ISM, the different roles they play, and the help and support they need from health care providers (HCPs)

Methods

- Two virtual ISM Caregiver Advisory Board meetings were conducted, designed to capture qualitative insights on experiential knowledge that complements clinical evidence, focusing on psychological burden, system navigation, and definitions of meaningful support and care in ISM

Study design

- 2x 4-hour Virtual Caregiver Advisory Boards
- June 27 and July 22, 2025[a]
- Caregivers (N=12) of patients with ISM
- Qualitative insights captured

Advisory board objectives

- Characterize the lived experience of caregivers supporting patients with ISM
- Define caregiver roles across diagnosis, disease management, and crisis preparedness
- Identify unmet needs and opportunities to better integrate caregiver insight into care delivery and engagement models

Outputs

- Recommendations for embedding caregiver perspectives into patient engagement and care strategies

[a]The two advisory boards were conducted as separate sessions to better accommodate the time constraints of caregiver advisors.
ISM, indolent systemic mastocytosis.

Caregiver demographics

Number of caregivers

N=12

Who caregivers provided care for

- Caregivers for their spouses
- Caregivers for a parent or child

Number of years to diagnosis for the individual with ISM

[a]Specific time not defined/captured.

Overview of Insights from Caregivers

- Caregivers described profound emotional exhaustion driven by prolonged diagnostic delays shaped by uncertainty, limited disease awareness, and repeated efforts to validate symptom complexity
- Many reported social isolation, burden of sustained crisis readiness, and ongoing role strain as they moved between identities of partner, nurse, navigator, and advocate, often without formal recognition or support
- Clinical interactions frequently required caregivers to re-center conversations on the person living with ISM to preserve autonomy and ensure their lived experience remained visible
- Frustration stemmed from fragmented care, lack of shared terminology, limited symptom awareness/recognition by HCPs, and misaligned definitions of disease "control"
- Unpredictability was identified by all caregivers as the most difficult aspect of ISM, driving constant vigilance, crisis preparedness, and long-term emotional strain
- In a disease defined by symptom variability, unpredictability shapes caregiver behavior, care coordination needs, and perceptions of control
- Many caregivers expressed an urgent need for greater support and resources to help them care for their loved ones
- Despite these pressures, caregivers demonstrated resilience and emphasized the importance of peer connection and proactive advocacy, underscoring the need for care models that recognize and support the caregiver role

Diagnostic uncertainty creates chronic emotional and cognitive load

Diagnostic uncertainty and prolonged time to recognition create sustained strain for caregivers and people living with ISM

- Caregivers described sharing the full emotional burden of the diagnostic journey, particularly when symptoms were fluctuating, atypical, or difficult to contextualize clinically
- Caregivers emphasized that clinical encounters were most productive when discussions remained centered on their loved one's lived experience and functional impact

"Both symptoms and disease are important…symptoms are right there in front of you. Disease is way under the hood…even if there's a 0.1% chance that it's a certain disease, it's there."

Male, spouse of an individual living with ISM

Caregivers as care partners:
- Acknowledge caregiver observations as longitudinal context that can support clinical decision-making
- Utilize a model of care management that is multifaceted and truly reflects the experiences and needs of caregivers and their loved ones

Healthcare team interactions: variability in care experience

- Caregivers described variability in healthcare team interactions, noting that visits were most challenging when care focused narrowly on individual symptoms rather than their loved one's broader lived experience
- Caregivers appreciate a medical team that brings both deep clinical expertise and a holistic view of their loved one's physical and emotional well-being
- When visits focus on short-term clinical status, caregivers may feel the cumulative impact of symptoms is not fully captured

"This disease has robbed my husband of things he used to enjoy. Just because you can't see it or his 'labs are normal' doesn't mean what he's experiencing isn't real."

Female, spouse of an individual living with ISM

From listening to leveraging caregiver insight:
- Encourage caregivers to attend appointments for people living with ISM, given the important role that caregivers play, while continuing to keep the focus on the person living with ISM
- Consider relational, not just clinical, competencies when delivering care; treat caregiver observations as critical data, not background noise
- When approaching diagnosis, understand that individuals may not have a "typical" presentation and that caregivers can provide valuable insights

Clinical stability does not equal lived stability

Misaligned definitions of disease control between caregivers/people living with ISM, and healthcare professionals

- People living with ISM may appear to be clinically stable over time, but may experience persistent, unpredictable symptoms
- Caregivers would define disease control as predictability, reduced flares, and ability to function, emphasizing that baseline stability does not necessarily reflect daily functioning or quality of life

"No progression doesn't mean good quality of life. It means we're stuck in survival mode."

Female, spouse of an individual living with ISM

Aligning care with real-world outcomes:
- Redefine treatment success beyond no worsening of symptoms; place equal value on stability, functionality, and improved quality of life
- Incorporate shared language around symptom burden and daily functioning into care conversations

The human cost of carrying these burdens

Isolation takes an emotional toll

- Creeping isolation and/or loneliness compounds the physical and emotional toll
- Caregiver consensus was that nobody in their circle truly understands what they're going through

"Lonely is a word I'm a little too comfortable with... We've lost most of our friends."

Female, spouse of an individual living with ISM

Keeping caregivers connected:
- Ensure that caregivers are aware of available resources to address their isolation, such as support groups

Self-sacrifice outweighs self-care and leads to burnout

- Caregivers admitted they don't get much time to themselves. Their time off is filled with medical appointments or hospital visits; many arrange their schedules around maintaining their loved ones' lives
- Caregivers understood they couldn't take proper care of their loved one if they were not taking care of themselves, but found self-care difficult to fit in or access

"It's hard to be supportive if you're not taking care of yourself. I've worked to restructure those things that reduce my stress and provide joy so I'm better able to support her."

Male, spouse of an individual living with ISM

Making caregiver support visible and accessible:
- Ensure caregivers are routinely made aware of credible, disease-relevant support resources so they are not left to manage the emotional and physical demands of ISM alone

Caregivers want more peer connection and a central hub for monitoring and support resources

- Caregivers expressed a desire for improved community connections and a release valve
- There was a desire to be able to connect with people that truly understand and can empathize with their situation

"It's really hard and overwhelming to do it all yourself. I'm struggling mentally."

Female, spouse of an individual living with ISM

Multi-faceted support for caregivers:
- Support for caregivers must include both emotional and practical strategies, such as support groups and resources for disease monitoring and management

Fragmented care creates invisible labor

Caregivers have multiple roles and responsibilities, contributing to identity strain

- Caregivers play a central, yet often unrecognized, role in supporting their loved ones and require resources to empower and optimize their efforts
- Caregivers do far more than support: they prep for visits, track symptoms, manage logistics, translate medical-speak, juggle daily demands due to the unpredictability of symptoms, and carry the emotional weight of social and family interactions
- Many caregivers described assuming advocacy roles to help bridge gaps across care settings and convey symptom patterns over time

"I am seen as both wife and caregiver. I often say 'as a caregiver' or 'as a wife' at the doctor to give both perspectives. When I say 'caregiver' I put my logical cap on. As a wife, I'm more emotional, caring, and loving."

Female, spouse of an individual living with ISM

Enabling shared decision-making with caregivers:
- Ensure understanding of caregiver roles in treatment decision-making, and how caregivers play their dual role so that shared decisions can effectively be made
- Ensure availability of resources for caregivers to help alleviate some of the burden of monitoring, management, and articulation of the unpredictability of ISM, to empower them to optimize and sustain their role in advocating for their loved ones

Summary

- Caregiver perspectives provide essential clinical context for care of people living with ISM
- Clinical stability may coexist with substantial daily burden that may not be fully captured during routine visits
- Unpredictability drives constant vigilance and care coordination
- Integrating caregiver observations as longitudinal context can support more sustainable disease management centered on people living with ISM
- Addressing caregiver needs is not ancillary, but foundational to effective care for people living with ISM

Conclusions

- Integrating caregiver observations as longitudinal context can strengthen communication, improve continuity of care, and support shared understanding of symptom burden and functional impact in complex, symptom-driven diseases such as ISM
- Effectively supporting caregivers in their role can improve the lived experiences and outcomes of people living with ISM

Acknowledgements

The authors thank the participants of the advisory board, which was funded by Blueprint Medicines Corporation, a wholly-owned subsidiary of Sanofi and managed by Lumanity Inc.

Medical writing support was provided by Ramya Kollipara, PhD, and editorial support was provided by Travis Taylor, BA, of the Prime Group of Companies (Knutsford, UK), and was funded by Blueprint Medicines Corporation, a wholly-owned subsidiary of Sanofi. Responsibility for all opinions, conclusions, and data interpretation lies with the authors.

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Conflicts of interest/disclosures

Dr Kurtin has no conflict of interest to disclose. For full author disclosures, please contact medinfo@blueprintmedicines.com.

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Presented at the American Academy of Allergy, Asthma & Immunology (AAAAI) 2026 Annual Meeting, February 27–March 2, in Philadelphia, PA, USA. Please contact medinfo@blueprintmedicines.com for permission to reprint and/or distribute.