Poster Number 514

Understanding Disease Control: What Shapes Patient Experience in Indolent Systemic Mastocytosis

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Introduction

- Systemic mastocytosis (SM) is a clonal mast cell disease driven by the *KIT* D816V mutation in ~95% of adult cases[1–4]
- Approximately 85% of individuals diagnosed with SM have indolent systemic mastocytosis (ISM), a form of the disease marked by unpredictable, often debilitating cutaneous, gastrointestinal, and systemic symptoms that can be life altering and significantly disrupt daily life.[5,6]
- When symptoms are not adequately controlled, people living with ISM report meaningful declines in quality of life, alongside increased healthcare utilization and a persistent burden that shapes their sense of stability and well-being[7,8]
- A disconnect persists between clinical and patient perspectives on disease control. Clinicians may consider ISM symptoms to be "well-controlled," yet patient-reported experiences often reveal a more nuanced reality. While clinicians aim to address the heterogeneous manifestations of ISM, they may not always recognize which symptoms patients find most disruptive. As a result, key issues may be insufficiently monitored or treated, leaving patients feeling sub-optimally controlled[9–11]
- Notably, 78% of advanced practitioner (AP) respondents reported that disease was well controlled in ≥50% of their patients; however, 54% of APs reported difficulty tracking symptom severity and burden over time[9]
- This treatment-agnostic survey analysis evaluated independent patient responses to identify factors that may influence patients' perceptions of disease control, providing insights to inform clinical care

Methods

- Original full survey[9]:
  - In collaboration with The Mast Cell Disease Society and the Advanced Practitioner Society of Hematology and Oncology (APSHO), an ISM patient survey and AP survey were developed by an AP-led steering committee incorporating validated tools to measure symptom burden, symptom burden impact, barriers, and strategies for improving symptom burden, and was conducted by APSHO
  - Treatment-agnostic surveys were embedded in Qualtrics and were deployed by Conexiant to a convenience sample of AP members of APSHO, AP members of the American Initiative in Mast Cell Diseases, and patients affiliated with The Mast Cell Disease Society between December 22, 2024, and February 3, 2025
- In this sub analysis, responses from patients with ISM were analyzed regarding symptom profile, support and care coordination, and self-management identity. Data were stratified by patient perceptions of well-controlled versus not optimally controlled disease based on the self-reported disease, control rating

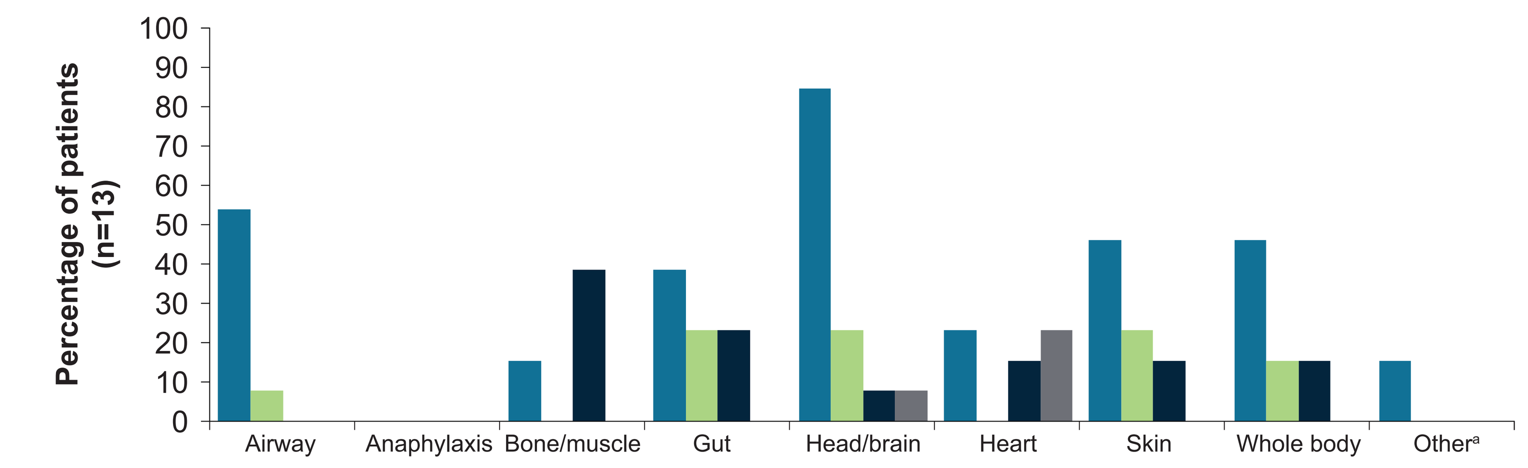
Results: ISM patient survey

Patient demographics among survey respondents (n=53)

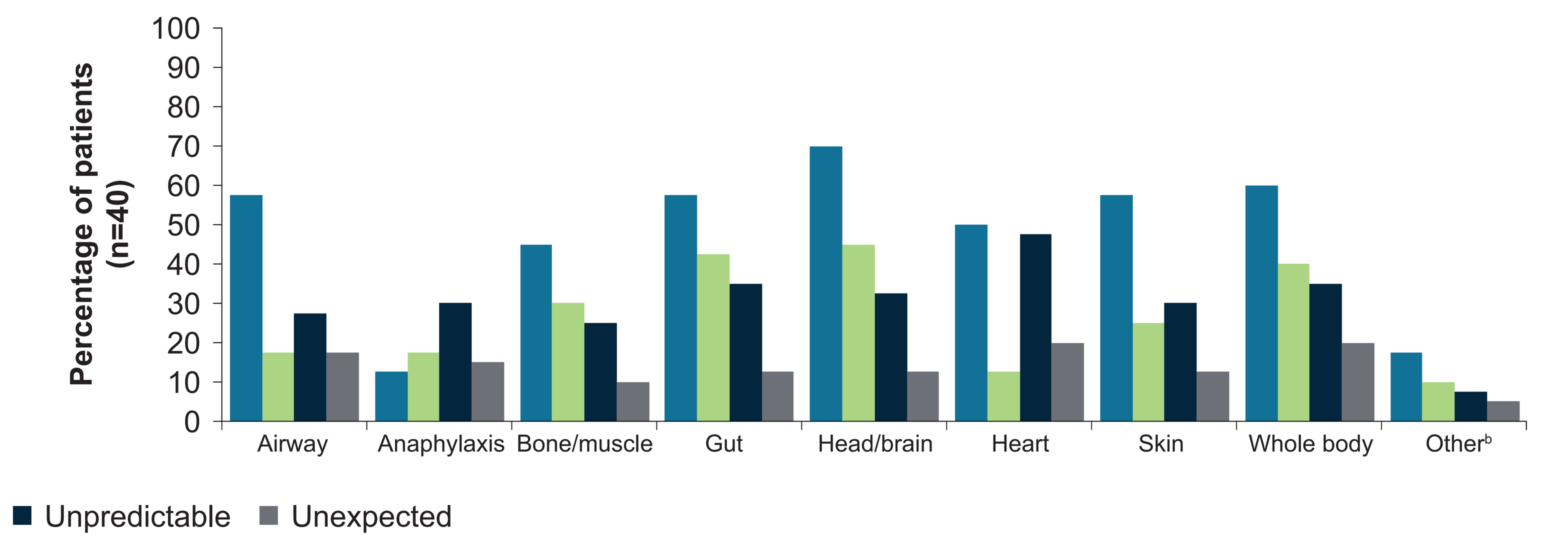
Mean age at ISM diagnosis, years (range)	44.5 (24–69)
Mean years since ISM diagnosis (range)	8 (<1–43)
Time from symptom onset to confirmed diagnosis, n (%)	
<1 year	8 (15)
1–3 years	6 (11)
4–7 years	12 (23)
8–10 years	7 (13)
>10 years	20 (38)
Patients who consider their disease to be well controlled, n (%)	
Yes	13 (25)
No	40 (75)

Not optimally controlled ISM was associated with greater symptom frequency, severity, and unpredictability

Patients reporting well-controlled disease

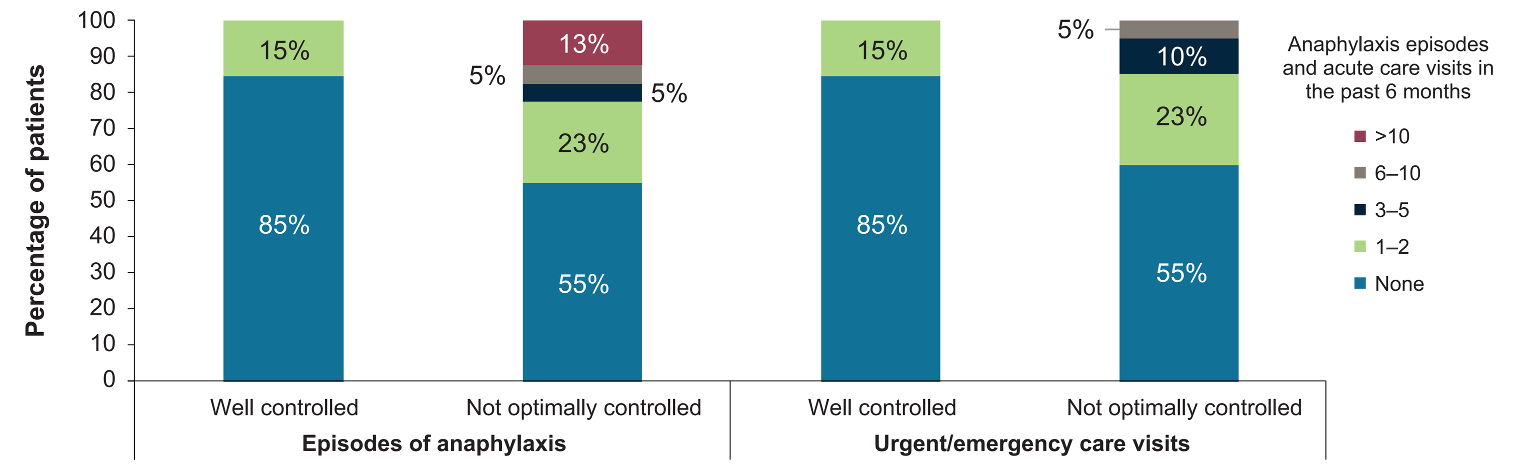


Patients reporting not optimally controlled disease

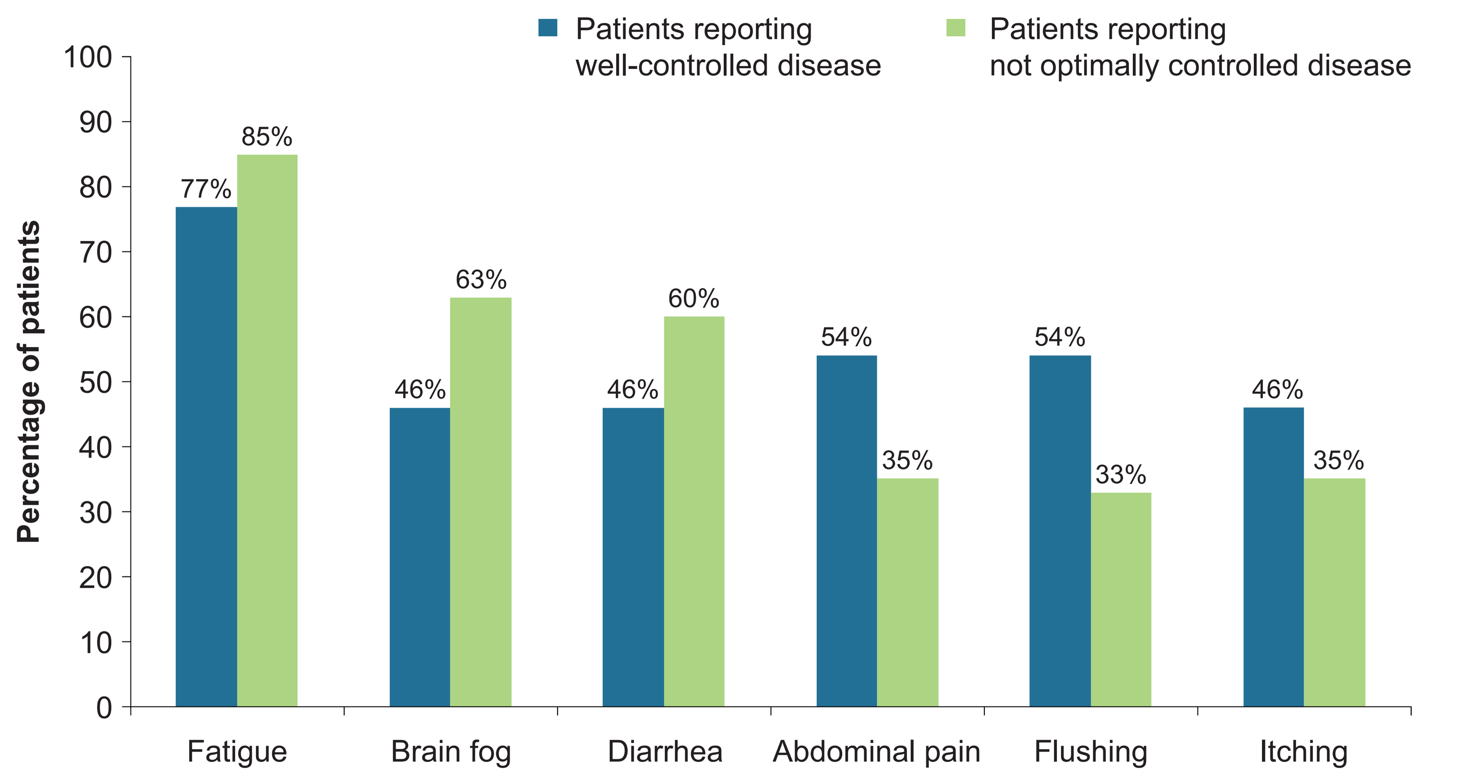


[a]Includes anxiety and indigestion/heartburn. [b]Includes osteoarthritis, odor sensitivity, inflammation, exacerbation of other chronic illnesses, joint pain, bloating, mood swings, and unpredictability of symptoms.

Patients with not optimally controlled ISM reported higher rates of anaphylaxis and urgent or emergency care visits

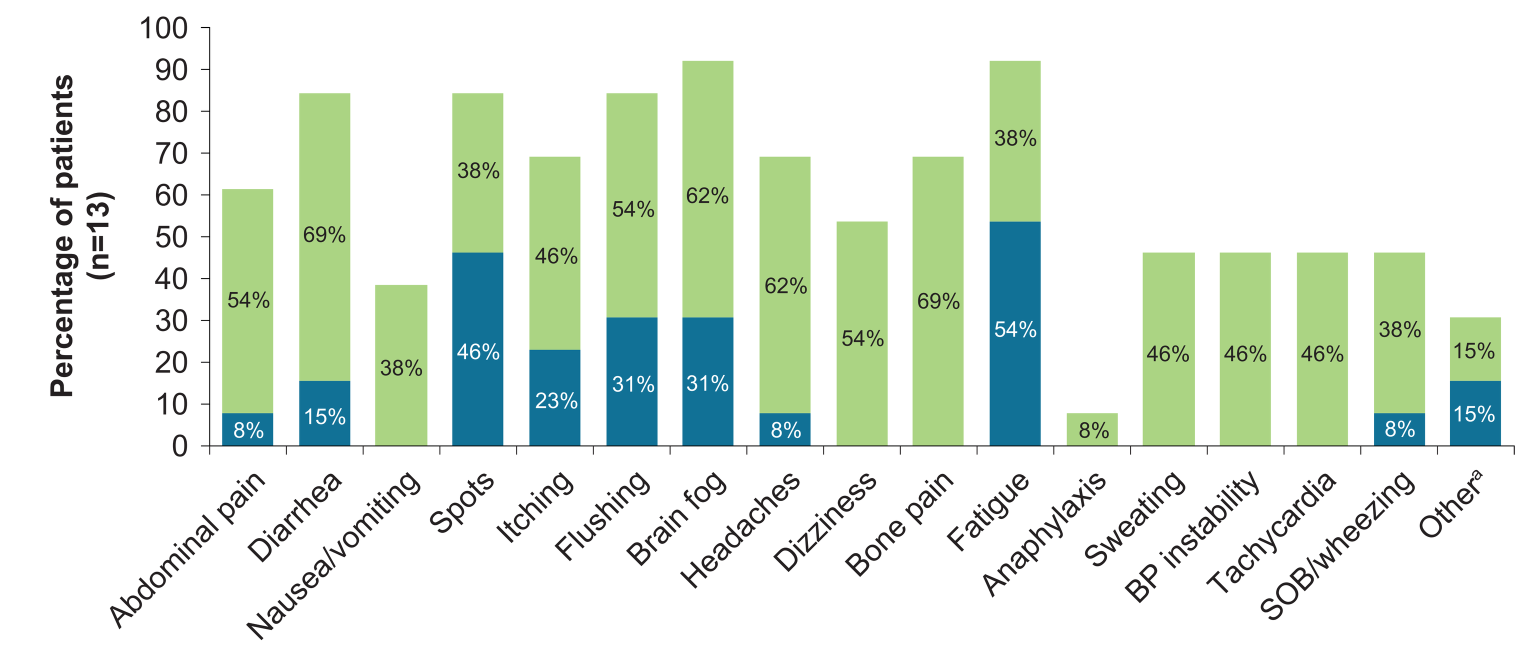


Even with well-controlled ISM, patients reported similar symptoms, though at lower rates than those reporting not optimally controlled disease

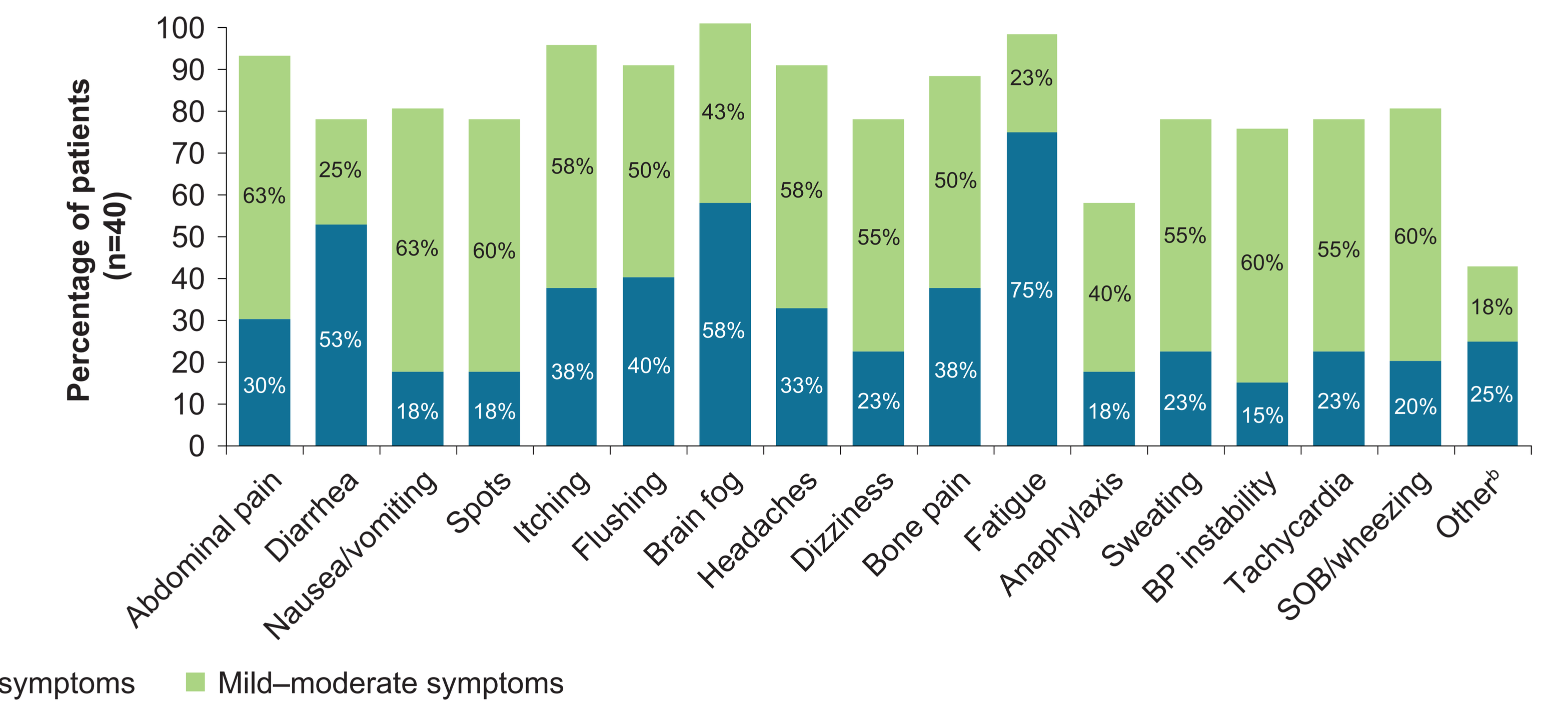


Severe symptoms were reported even in well-controlled disease, despite overall reduced frequency of severe symptoms compared with patients reporting not optimally controlled disease

Patients reporting well-controlled disease



Patients reporting not optimally controlled disease



[a]Includes anxiety, chest tightness, cold sores, and indigestion/heart burn. [b]Includes skin sensitivity, joint pain, and muscle aches.
BP, blood pressure; SOB, shortness of breath.

Conclusions

- Even when patients believe their disease is controlled, many still experience a meaningful burden that affects their daily lives
- Perceived disease control did not greatly change which symptoms most affected quality of life, with fatigue, cognitive, gastrointestinal, and dermatologic symptoms remaining prominent
- Not optimally controlled ISM was associated with greater symptom frequency, severity, unpredictability, and healthcare utilization, including anaphylaxis and urgent or emergency care visits
- These findings highlight the heterogeneous and patient-reported impacts of ISM on symptoms and quality of life, supporting the need for more patient-centered approaches to defining and monitoring disease control

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Conflicts of interest/Disclosures

Dr Kurtin has no conflict of interest to disclose. Full disclosures for all authors are available upon request at medinfo@blueprintmedicines.com.

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