Patient Perceptions in Mast Cell Disorders

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KEYWORDS

• Mast cell disorder • Mast cell activation • Patient perceptions and experiences
• The Mastocytosis Society • Anaphylaxis • Quality of life • Support • Disability

KEY POINTS

- A wide range in frequency and intensity of mast cell activation symptoms exists among individual mast cell disorder patients and also collectively in this population.
- Mast cell disorder patients report both disruption and reduced quality of life, with possible financial repercussions, due to physical and/or neuropsychiatric symptoms, including anaphylaxis, and their unpredictable onset.
- Triggers of mast cell activation, some of which may be less recognized than others, vary widely and can include heat/cold, stress, fatigue, foods/beverages, alcohol, medications/contrast, venoms, odors, infections, and exercise.
- Patients report that treatment of mast cell disorders is primarily directed at symptom reduction rather than cure in all but the most advanced variants.

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INTRODUCTION

The ability of health care professionals and industry representatives to understand the experiences, perceptions, and perspectives of patients plays a vital role in successful care, treatment, and informed development of novel therapies. Regulators increasingly recognize the patient's voice as critical to drug development, with the US Food and Drug Administration's 2009 Guidance on Patient-reported Outcomes, patient-focused drug development meetings, and the 21st Century Cures Act. Patients with a mast cell disorder (MCD), including mastocytosis, mast cell activation syndromes (MCAS), and hereditary α-tryptasemia, may experience daily physical, emotional, and social stressors, and awareness of these factors can help medical professionals provide more comprehensive care. Recognition that patient perceptions of their illness may differ from perceptions of treating physicians, especially related to quality of life, degree of disability, and chronicity of symptoms, is essential.

PATIENT POPULATION AND CHARACTERISTICS

MCDs, considered rare diseases, affect newborns to adults and are divided into clonal and nonclonal disorders. Clonal disorders include cutaneous mastocytosis (CM), systemic mastocytosis (SM), and monoclonal MCAS.[1–6] Nonclonal disorders include forms of MCAS that are secondary or idiopathic.[1,2,7] CM often presents in children less than 2 years old, and variants, most commonly maculopapular CM (urticaria pigmentosa), differ in size, shape, and pattern of skin rash, percentage of skin affected, and frequency of persistence into adulthood.[8] Cutaneous mastocytoma or mastocytomas, usually present at birth, may spontaneously regress during childhood.[3,5,8] SM, which may be associated with cutaneous lesions, is divided into indolent SM (ISM), smoldering SM (SSM), and the more advanced SM (AdvSM) categories: SM with associated hematologic neoplasm (SM-AHN), aggressive SM (ASM), and mast cell leukemia (MCL).[3–6] A separate category exists for mast cell (MC) sarcoma.[3,5] Progression from indolent to more aggressive disease is defined by strict diagnostic criteria.[3–6] A more recently described entity, hereditary α-tryptasemia, is associated with MC mediator release symptoms, dysautonomia, and connective tissue disorders, especially joint hypermobility.[9] Potential triggers of MC mediator release,[1,10,11] and resulting symptoms,[2,7,11,12] vary for each MCD patient (**Boxes 1** and **2**). Patients

Box 1
Common triggers for patients with a mast cell disorder

- Heat, cold, sudden temperature changes
- Emotional, physical, environmental stress
- Fatigue
- Exercise, friction, vibration, surgery
- Food, beverages, including alcohol
- Medications (opioids, NSAIDs, antibiotics, and anesthetics)/contrast dyes
- Hymenoptera venom
- Odors
- Infections

Data from Refs.[1,10,11]