Unravelling the Challenges of MCAS Diagnosis and Management: Insights from a UK Patient Survey

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INTRODUCTION

Mast Cell Activation Syndrome (MCAS) is a condition in which abnormal mast cell activation results in a wide array of symptoms affecting multiple organ systems.[1] The variability and non-specific nature of symptoms often leads to diagnostic delays.[2] Many individuals with MCAS present with comorbid conditions such as Ehlers-Danlos Syndrome (EDS), Hypermobility Spectrum disorders and Postural Orthostatic Tachycardia Syndrome (POTS), further complicating the diagnostic process.[3] Additionally, treatment is often inconsistent, with many patients relying on more dynamic approaches to manage their symptoms.[2,3]

OBJECTIVE

To explore the experiences of individuals living with confirmed or suspected MCAS in the UK, including symptom burden, diagnostic process, treatment, and overall experiences of care.

METHODOLOGY

A survey was distributed in January 2024 to people across the UK with confirmed or suspected MCAS. Patients were recruited through a combination of social media outreach, support groups, and community networks to ensure a diverse and representative sample.

RESULTS

The survey was completed by 130 people, mostly online (n=107). Survey respondents were predominantly adult (97%), female (94%) and White British (88%, Figure 1). Of 130 respondents, 87 had a confirmed MCAS diagnosis, while 43 were undiagnosed (Figure 2). Most respondents (97%) had symptoms consistent with MCAS for 2 years or more, and 19 undiagnosed patients had been experiencing symptoms for more than 11 years (Figure 3). The most common comorbidities were EDS/HMS (51 cases), POTS (41 cases), and fibromyalgia (33 cases).

Figure 1: Ethnicity

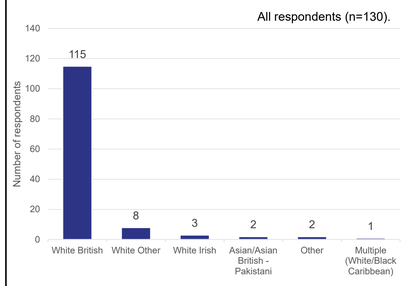


Figure 2: MCAS diagnosis status

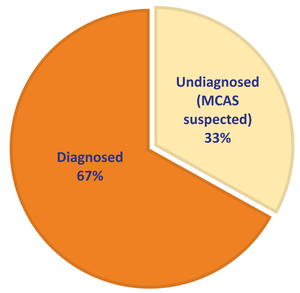


Figure 3: Time since first symptom onset

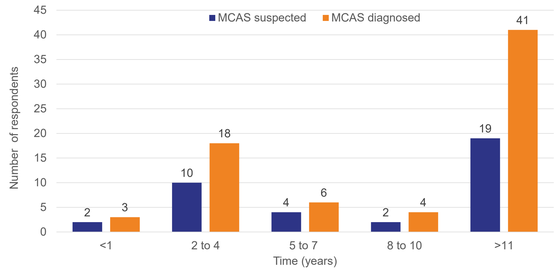


Figure 4: Age at diagnosis

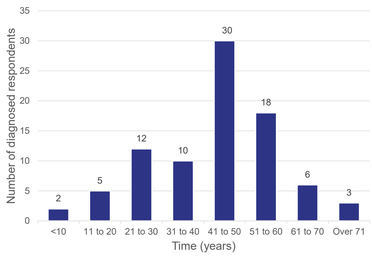


Figure 5: Time since MCAS diagnosis

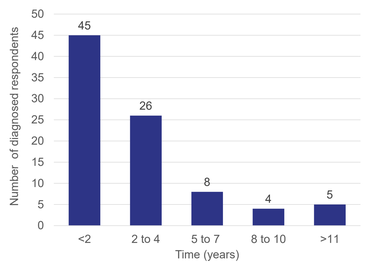
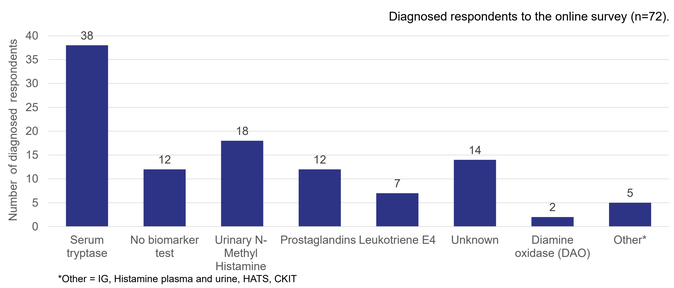
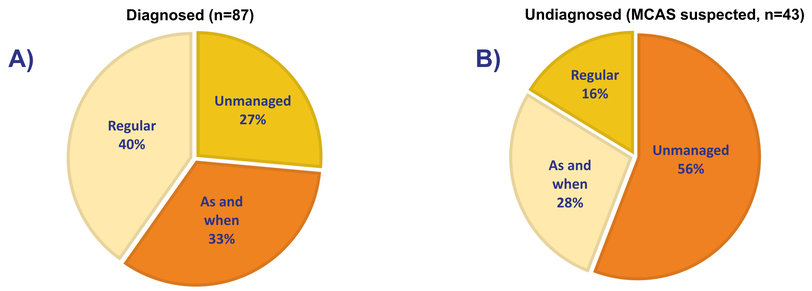


Figure 6: Biomarker tests to investigate MCAS diagnosis



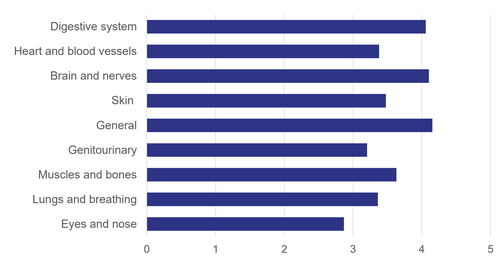
*Other = IG, Histamine plasma and urine, HATS, CKIT

Figure 7: Access to care for MCAS (diagnosed vs undiagnosed)



Based on responses to the question "How is your MCAS currently managed? For example, you may be seen as and when symptoms arise, regularly seen under the pain management team or you may email/speak with your medical team as needed."

Figure 8: Symptom burden in past 3 months



Average rating for "how bothersome were these symptoms" (0= least and 5=most). Diagnosed respondents to the online survey (n=72).

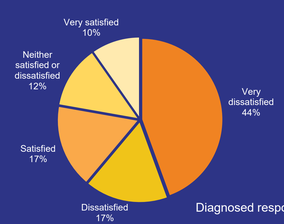
Patient experiences and satisfaction

"Horrific. Affects every part of my life."

"When having emergency admissions to hospital, I was often not believed/not treated with the correct medication."

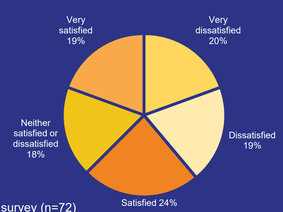
"Until I was diagnosed it was devastating. Now I can control it I can live an almost normal life."

Satisfaction with overall diagnostic process



Diagnosed respondents to the online survey (n=72)

Satisfaction with overall MCAS treatment and management plan



DIAGNOSTIC PROCESS

Among respondents with a confirmed MCAS diagnosis, average age at diagnosis was 43.1 (range 2 to 77 years, Figure 4), and most (51%) had received an MCAS diagnosis within the past 2 years (Figure 5). Almost half of diagnoses (46%) were made by allergists or immunologists.

Although serum tryptase elevation was the most common biomarker test performed (Figure 6), all reported serum tryptase levels were within normal range and therefore did not positively contribute to MCAS diagnosis. Twelve people reported confirmed MCAS diagnosis based on symptoms and/or response to therapy alone, with no biomarker tests performed.

TREATMENT & MANAGEMENT

Among respondents with confirmed MCAS diagnosis, 40% reported regular access to care for their condition, while 27% said that their MCAS was unmanaged, and 33% received care as needed (Figure 7). Overall, 98 survey respondents (75%) reported that they were receiving treatment for MCAS. Treatments included prescribed and over-the counter medications, vitamins and supplements. The main drug classes used during the course of MCAS treatment were antihistamines (H1 antagonists: fexofenadine, cetirizine, ketotifen, loratadine, chlorphenamine; H2 antagonists: famotidine). Mast cell stabilisers (sodium cromoglicate capsules or eyedrops) were also commonly used.

IMPACT OF SYMPTOMS

Diagnosed respondents reported symptoms across multiple organ systems within the 3 months prior to completing the questionnaire (Figure 8). On average, neurological symptoms were among the most burdensome (for example, headaches, brain fog, numbness, depression and anxiety), as well as digestive symptoms, and general symptoms such as extreme tiredness, chills, overheating, trouble sleeping, and anaphylaxis.

PATIENT EXPERIENCE

MCAS has a profound impact on respondents' lives, with some facing disbelief from healthcare professionals and inadequate treatment during emergencies. Many respondents were dissatisfied with the overall diagnostic process and with their MCAS treatment and management plan. Respondents stressed the need for better coordination of tests and increased awareness of MCAS among healthcare professionals. The lengthy and frustrating diagnosis process was a common theme, but once diagnosed and managed, some reported substantial symptom improvement.

CONCLUSION

Survey data from this predominantly White British adult female cohort illustrate the significant variation and challenges associated with MCAS diagnosis and management in the UK. Despite serum tryptase being an outdated and uninformative biomarker for MCAS, serum tryptase testing was commonly reported as part of diagnostic investigations. More than a quarter of respondents with a confirmed MCAS diagnosis reported that they did not have regular or as-needed access to a specialist. In conclusion, the data indicates an urgent need to standardise diagnosis and treatment pathways and expand specialist training in MCAS.

References

1. Frieri, M. (2018). Mast cell activation syndrome. Clinical reviews in allergy & immunology, 54, 353-365.
2. Castells, M., Giannetti, M. P., Hamilton, M. J., et al. (2024). Mast cell activation syndrome: Current understanding and research needs. Journal of Allergy and Clinical Immunology, 154(2), 255-263.
3. Jennings, S. V., Slee, V. M., Finnerty, C. C., et al. (2021). Symptoms of mast cell activation: the patient perspective. Annals of Allergy, Asthma & Immunology, 127(4), 407-409.

Abbreviations

DAO=Diamine Oxidase, EDS/HMS=Ehlers-Danlos Syndrome/Hypermobility Spectrum Disorders, HATS=Histamine N-Methyltransferase, IG=Immunoglobulin, H1, histamine receptor 1, H2=histamine receptor 2, MCAS=Mast Cell Activation Syndrome, ME/CFS=Myalgic Encephalomyelitis/Chronic Fatigue Syndrome, POTS=Postural Orthostatic Tachycardia Syndrome, UOE=Urinary Oxalate Excretion.

Acknowledgements

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