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Immune dysfunction, both mast cell activation disorders and primary immune deficiency, is common among patients with hypermobile spectrum disorder (HSD) or hypermobile type Ehlers Danlos Syndrome (hEDS)

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BACKGROUND

Manifestations of immune dysfunction has been reported to be a common in patients with hypermobile spectrum disorder (HSD) or hypermobile type Ehlers Danlos Syndrome (hEDS). The purpose of this study was to characterize the spectrum immune system dysregulation in patients with hEDS, which can range from primary immune-deficiency disorders (PID) to hypersensitivity syndromes, including mast cell activation syndrome (MCAS).

METHODS

This was a retrospective study of patients referred to a community-based allergy/immunology practice for evaluation of possible mast cell activation syndrome (MCAS). Immunological test results included mast cells (MC) activation markers (tryptase, histamine, prostaglandin metabolites) and an analysis of immunoglobulins, complement and MBL. Past medical histories and laboratory findings were analyzed.

RESULTS

Data from patients evaluated between from 2014 to 2019 was analyzed. Of the 986 patients, who underwent evaluation for mast cell disorders, 168 patients were found to have had a combination of either MCAS, HSD/hEDS or hypogammglobulinemia, and a combination of at least two of the three diagnoses. Further analysis identified the following combinations:

(1) 72/168 (42%) had HSD/hEDS and MCAS,
(2) 40 /168 (24%) had hypogammaglobulinemia and MCAS,
(3) 32/168 (20%) had hypogammaglobulinemia and HSD/ hEDS; and
(4) 24/168 (14%) had a combination of all three co-aggregating disorders.

Moreover, these observations reveal that 76% of HSD/hEDS patients presented with an immune co-morbidity, either PID or MCAS.

CONCLUSIONS

This study highlights the presence of immune dysfunction in patients with HSD/hEDS, either MCAS, PID or both; and these observations indicate the need to not only screen for MCAS inpatients with HSD/hEDS, but PID, which can present with features

immediate hypersensitivity. This, in turn, will impact therapeutic approaches for an EDS patient, who presents with immunological co-morbidities. Noting that fatigue and pain are common consequences of immune dysfunction, we propose that screening for both MCAS and PID should be part of evaluation of the initial screening of all HSD/ hEDS patients.

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DECLARATIONS OF INTEREST

Anne Maitland: Speakers' Bureau of Sanofi/Regeneron and Genentech/Novartis