Hypermobility Spectrum Digestive Disorder (HSDD): An Underdiagnosed Cause of Chronic GI Symptoms

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Introduction

Hypermobility spectrum disorder (HSD) is a connective tissue disorder characterized by generalized joint hypermobility and variable systemic involvement. Beyond musculoskeletal complaints, patients may experience gastrointestinal, autonomic, dermatologic, and neurologic symptoms. **Mast cell activation syndrome (MCAS)** is a frequently associated comorbidity, often contributing to multisystem symptoms including flushing, abdominal discomfort, and fatigue. Due to its broad clinical presentation, HSD is frequently underrecognized, leading to misdiagnosis and delayed treatment.

Literature Review

- Individuals with hypermobility syndromes frequently experience gastroesophageal reflux, bloating, dysmotility, and food intolerances, often accompanied by autonomic disturbances such as dizziness and palpitations [1]. This guidance emphasizes the need for a comprehensive, multisystem approach to evaluation and management.
- HSD remains underdiagnosed due to lack of clinician familiarity and experience with the disorders, a highly variable clinical presentation, and absence of a confirmatory test [2].
- There is a frequent co-occurrence of MCAS in patients with HSD. Shared features include histamine-mediated symptoms such as GI distress, flushing, and fatigue, with many patients demonstrating significant symptom relief from dual H1 and H2 blockade [3].

Case Presentation

- **History of present illness (HPI):** 29-year-old South Asian male with chronic bloating since childhood, associated with acid reflux, constipation with straining, and insomnia.
- **Review of symptoms:** positive for anxiety, snoring, dizziness, fatigue, trouble speaking, back pain, nasal congestion, and postnasal drip
- **Past medical history:** multiple ligament/tendon injuries, right shoulder tendon rupture, childhood ankle fracture from a fall; EGD 8 years ago revealed H. pylori with duodenal erosion which was treated
- **Social history:** married, works in IT, active with yoga, weightlifting, and constant exercise
- **Physical exam:** Beighton score 9/9
  - 1 point: forward flexion of trunk with palms flat on floor
  - 2 points: hyperextension of right and left knees >10°
  - 2 points: passive apposition of right and left thumbs to forearm
  - 2 points: Passive dorsiflexion of right and left fifth fingers >90°
  - 2 points: hyperextension of right and left elbows >10°

Physical Exam Findings

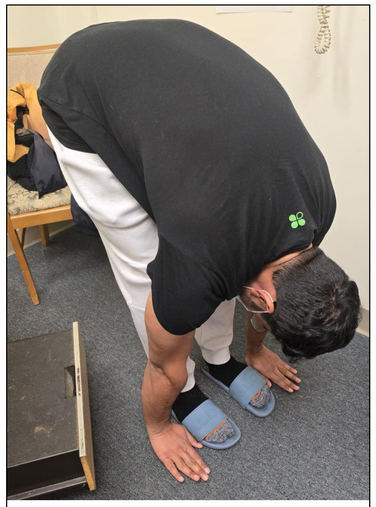

Fig. 1: Forward flexion with palms flat

Fig. 2: Knee hyperextension

Fig. 3: Thumb to forearm

Fig. 4: Pinky finger passive dorsiflexion

Fig. 5: Elbow hyperextension

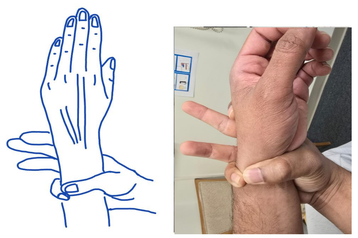

Fig. 6: Positive wrist (Walker-Murdoch) sign

Fig. 7: Reverse prayer sign

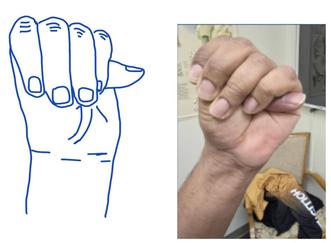

Fig. 8: Positive thumb (Steinberg) sign

Unique Aspects of the Case

- **Delayed recognition:** HSD was not considered until a comprehensive evaluation highlighted multisystem involvement and prompted further screening.
- **Misdiagnosis risk:** Initially labeled as irritable bowel syndrome (IBS), the patient exemplifies how HSD can mimic common conditions, leading to prolonged ineffective treatment.
- **Connective tissue fragility:** History of multiple ligament/tendon injuries and childhood fractures along with generalized joint laxity (Beighton score 9/9) supported a systemic, lifelong connective tissue disorder.
- **Overlap with MCAS:** Symptomatic improvement with H1/H2 blockade underscores the importance of considering coexisting MCAS in HSD patients.

Conclusion

This case demonstrates the clinical importance of considering HSD in patients with chronic, treatment-resistant gastrointestinal symptoms. Recognition of overlapping conditions like MCAS allows for more precise therapy; in this case, targeted histamine blockade with famotidine and loratadine led to substantial symptom relief. Greater awareness, coupled with the use of the Beighton scoring system as a simple bedside screening tool, can aid in identifying joint hypermobility, facilitating timely diagnosis and guiding appropriate management.

Recommendations

- **Suspect HSD and MCAS** in patients with chronic, multisystem complaints.
- Take a **thorough history and physical**, including **Beighton screening**.
- Consider adopting the proposed term **"hypermobility spectrum digestive disorder (HSDD)"** to more accurately describe this distinct GI phenotype associated with HSD.
- Promote **early, multidisciplinary recognition** to avoid diagnostic delays, enable more targeted treatment, and significantly improve patient outcomes.

References

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