Psychiatric evaluation and management of malnutrition in patients with hypermobile Ehlers-Danlos syndrome (hEDS)

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Background

- **What is hypermobile Ehlers-Danlos Syndrome (hEDS)?**
  - A collagen production disorder characterized by joint hypermobility with non-specific physical exam findings that is commonly associated with gastrointestinal (GI) symptoms[1]. hEDS is a clinical diagnosis.
  - It is the most common subtype of EDS, but it does not have an associated genetic marker or known mechanism of pathophysiology[1].
- **Psychiatric comorbidities are common:**
  - hEDS is associated with a higher incidence of anxiety disorders, depression and eating disorders as well as heightened somatization[2,3].
  - Symptoms in common with restrictive eating disorders include GI dysmotility, food avoidance/intolerance and autonomic instability[4].
  - It has been hypothesized that these symptoms may play a role in the development of nutrition-avoidant or restrictive eating habits in patients with hEDS[4].
- **Diagnostic & therapeutic challenges:**
  - It is difficult to distinguish organic from psychosomatic pathology.
  - Psychiatric symptoms are challenging to differentiate and can include a combination of the following:
    - somatic symptoms with conditioned avoidance behaviors
    - the development of a comorbid eating disorder (ED)
    - surreptitious illness exacerbation for psychological or other secondary gain
  - Weight restoration can become exceedingly complex when physiological limitations to enteral feeding are present in the context of GI dysfunction, pathological avoidance and/or intentional food restriction.
  - Diagnosis and treatment planning are complicated by the involvement of multiple disciplines or even institutions, leading to missed opportunities to achieve diagnostic clarity and prevent unnecessary intervention[5].

2017 Consensus Diagnostic Criteria of hEDS[6,7]

Criterion 1	Criterion 2	Criterion 3
1 of the following: ❑ Current hypermobility as defined by Beighton Score (minimum is >4-6 depending on patient's age) ❑ History of hypermobility	2 of the following: ❑ 5 or more additional clinical features suggestive of hEDS (including but not limited to dermatologic, orthopedic or cardiac anomalies) ❑ Positive family history of hEDS ❑ Acute chronic pain, chronic widespread pain or recurrent, non-traumatic joint dislocations	All of the following: ❑ Lack of signs suggesting another EDS subtype ❑ Other connective tissue disorders, including autoimmune conditions have been ruled out ❑ Absence of another condition known to cause hypermobility

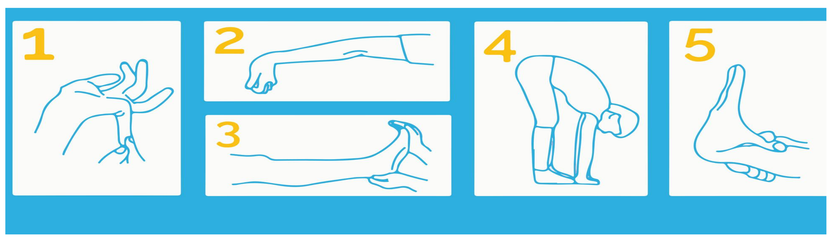
Patient cases & the role of the consulting psychiatrist

We present the cases of two adult patients with hEDS admitted for severe malnutrition with suspected contribution from an eating, functional GI and/or somatic symptom disorder. Psychiatric consultation was sought for evaluation, assistance with management of psychiatric comorbidities and guidance on feeding strategies. Each patient ultimately required multiple medical interventions including total parenteral nutrition (TPN), and their psychiatric comorbidities served as barriers to interdisciplinary agreement on the method of refeeding and delays in effective care. These cases demonstrate the diagnostic ambiguity and complexity in management of malnutrition in patients with reported or suspected hEDS.

Case #1

Case #2

Presenting signs & symptoms	Differential diagnosis	Interventions & complications
- PO intolerance - Malnutrition - Electrolyte derangements - Hypotension - Weight loss/Low body weight - Abdominal pain - Constipation - Multiple indeterminate diagnostic studies - History of abdominal surgeries without clear indication	**Psychiatric[2]:** - Avoidant-Restrictive Food Intake Disorder (ARFID) - Anorexia Nervosa (AN) - Somatic symptom disorder - Functional GI disorder - Factitious Disorder - Opioid Use Disorder **GI disorders associated with or sequelae of hEDS[8]:** - Gastritis/esophagitis - Hiatal hernia - Accelerated or delayed gastric emptying and/or colonic transit - Diverticulosis - Irritable bowel syndrome - Rectal evacuation disorders	- Long & short term use of total parenteral nutrition - Indwelling line placement with secondary infections and thrombosis - Chronic opioid therapy with development of physiologic dependence - Percutaneous feeding tube placement - Prolonged hospitalization - Repeated invasive diagnostic studies - Marked impairments in psychosocial functioning



Beighton Score Criteria[9]: Maneuvers tested on left & right for a total maximum score of 10

Discussion

Overlap with disordered eating:

Restrictive eating, whether secondary to an ED, somatic symptoms or an organic condition such as hEDS, can impair gut motility and lead to worsened abdominal discomfort. This can perpetuate ongoing food avoidance[10,11]. Common barriers exist in refeeding in patients with restrictive eating, regardless of the primary etiology. Evidence-based strategies for weight restoration in patients with restrictive EDs emphasize the importance of oral refeeding and recommend avoiding the use of tube feeds or parenteral nutrition whenever possible[12,13]. Psychological and behavioral treatments including exposure and response prevention, cognitive remediation therapy and other therapy modalities designed to increase distress tolerance or change maladaptive behaviors have demonstrated association with weight gain in patients with restrictive EDs[14,15,16]. These therapies target anxious, phobic and obsessive-compulsive thought patterns that are prominent in AN, ARFID, somatic symptom disorder, functional disorders, and potentially hEDS given the overlap with these conditions. This suggests that the role of psychotherapy should be explored further in the treatment of malnutrition in hEDS.

Other diagnostic considerations:

It is also important to discuss the possibility for misdiagnosis in hEDS. Symptoms are subjective and the presentation can be highly variable. There are no current consensus guidelines on management of hEDS, so clinical courses tend to be heterogeneous. Given that hEDS symptoms are generally subjective and nonspecific, this diagnosis allows susceptible patients to identify with an illness in order to meet subconscious psychological needs. Likewise, it offers the opportunity to consciously mimic illness for the purpose of secondary gain. These patients are at an elevated risk for iatrogenic harm, and this should be carefully considered before invasive interventions are pursued in patients with hEDS. There is no evidence to support the use of TPN or other surgical procedures to treat GI symptoms in this population. Multidisciplinary coordination is crucial for ensuring appropriate, effective, and compassionate care while preventing harm in this complex population.

Key points:

★Patients presenting with malnutrition in the context of hEDS should receive a thorough medical work-up and comprehensive psychiatric evaluation. A multidisciplinary approach should be taken in their care.

★The lack of genetic markers and clinical subjectivity may lead to misdiagnosis of hEDS. A differential diagnosis should include eating disorders and somatic symptom or functional disorders. Factitious disorder, substance use disorders and malingering should also be considered in appropriate cases.

★Psychotherapeutic techniques to manage unpleasant symptoms while reducing medicalization as a mechanism for coping with psychological distress may have a role in the treatment of hEDS. This warrants additional study.

★The diagnosis of hEDS should be confirmed and behavioral interventions should be exhausted before patients are subject to invasive procedures to treat malnutrition or other GI symptoms. Risks and benefits should be carefully weighed before pursuing TPN.

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