What is hypermobile Ehlers-Danlos Syndrome (hEDS)?

It is the most common subtype of EDS, but it does not have an associated somatic symptoms with conditioned avoidance behaviors.

Psychiatric comorbidities are common:

- It is difficult to distinguish organic from psychosomatic pathology.

Diagnostic & therapeutic challenges:

- Background

1 of the following:

- Positive family history of other connective tissue disorders
- Autonomic symptoms
- History of abdominal surgeries without clear indication

Criterion 1

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

- 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 diagnosis:

- 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:

- Gastritis/Esophagitis
- Hiatal hernia
- Accelerated or delayed gastric emptying and/or colonic transit
- Diverticulosis
- Irritable bowel syndrome
- Rectal evacuation disorders

Interventions & complications:

- Long & short term use of total parental nutrition
- Intravascular 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

2017 Consensus Diagnostic Criteria of hEDS

<table>
<thead>
<tr>
<th>Criterion 1</th>
<th>Criterion 2</th>
<th>Criterion 3</th>
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<tbody>
<tr>
<td>1 of the following:</td>
<td>2 of the following:</td>
<td>All of the following:</td>
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<tr>
<td>Current hypermobility as defined by Beighton Score (minimum is &gt;4-6 depending on patient’s age)</td>
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<td>2 of the following:</td>
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<tr>
<td>Positive family history of hEDS</td>
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<td>Lack of signs suggesting another EDS subtype</td>
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<td>Acute chronic pain, chronic widespread pain or recurrent, non-traumatic joint dislocations</td>
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<td>Other connective tissue disorders, including autoimmune conditions have been ruled out</td>
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<tr>
<td>Absence of another condition known to cause hypermobility</td>
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Beighton Score Criteria: Maneuvers tested on left & right for a total maximum score of 10

Malnutrition

PO intolerance

Gastritis/esophagitis

Functional GI disorder

Gastritis/esophagitis

Beighton Score

1. Current hypermobility as defined by Beighton Score (minimum is >4-6 depending on patient’s age)
2. Positive family history of hEDS
3. Acute chronic pain, chronic widespread pain or recurrent, non-traumatic joint dislocations

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. Common barriers exist in refueling 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 refueling and recommend avoiding the use of tube feeds or parenteral nutrition whenever possible.
- 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.
- 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.