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Hypermobile Ehlers Danlos Syndrome (hEDS)

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What is hEDS?

hEDS is an autosomal dominant disease with no known common genetic etiology that also exhibits locus heterogenicity and female sex dominance. It is part of the family of Ehlers Danlos Syndromes and is the most common Hereditary Disorder of Connective Tissue (HDCT) with an estimated population impact of 10 million in the US (Tinkle et al., 2017, pg. 49).

Diagnostic Criteria

It was previously known as:

- Ehlers–Danlos syndrome Type III
- Ehlers–Danlos syndrome Hypermobility Type (EDS-III / EDS-HT) and
- Joint Hypermobility Syndrome (JHS)

Updated diagnostic criteria was established at the 2017 International Consortium on Ehlers-Danlos Syndromes and Related Disorders and delineates hEDS at a specific point on a spectrum of joint hypermobility. Updated criteria aims to facilitate scholarship and understanding of specific genetic influence and disease process. hEDS diagnosis requires a combination of all parts of the following three categories:

1) Generalized Joint hypermobility (often per Beighton score)
2) Five or more classical features
3) Absence of positive alternative EDS or other disqualifying diagnosis. (The Ehers-Danlos Society, 2017).

Significance to Authorship

EDS is estimated to affect nearly 1 in 5000 individuals with hEDS including 80-90% of those diagnosed. hEDS presentation is found in the primary care setting, but despite this, the disease is poorly understood and underdiagnosed. This author's current family tree includes the genetic characteristics of hEDS. Personal experience with diagnosis and treatment proved difficult and prolonged as medical providers have a general lack of understanding of this disease and hesitate to treat the resulting complex patient needs. This scholarship is to improve awareness of hEDS to clinicians and improve delivery of care.

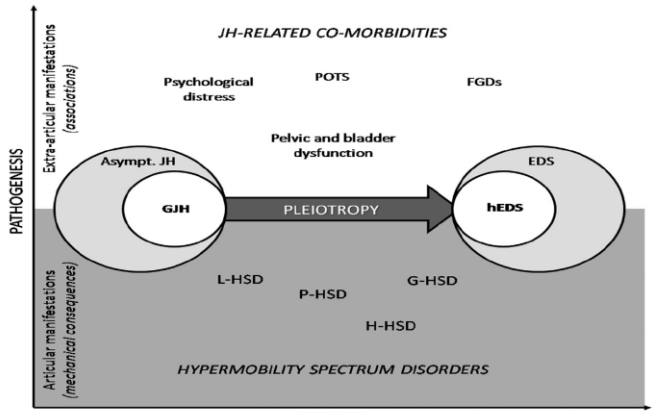

Figure 1. POTS- postural orthostatic tachycardia syndrome, FGDs- functional gastrointestinal disorders, JH- joint hypermobility, GJH- generalized joint hypermobility, L-HSD- localized hypermobility spectrum disorder, PHSD- peripheral hypermobility spectrum disorder (Castori et al., 2017)

Case Presentation

hEDS presents in 2 forms

1. Asymptomatic form- this form exhibits hypermobility but doesn't have the severity of pain and dysfunction
2. Symptomatic form- This form begins in childhood or early adolescence and progresses in severity of symptoms

3 phases of disease over lifetime

1. Hypermobile phase- presents in childhood. Usually seen with multiple dislocations and generalized gross and fine motor delays
2. Pain phase- 20-40 years of age. Headache, fatigue, pelvic pain often diagnosed as fibromyalgia
3. Stiffness Phase – joint trauma leads to arthritis, decreased muscle mass, weakness and disability (Tinkle et al., 2017).

Pathological Significance

hEDS is a disease which impacts almost every body system and exhibits varying degrees of significance in each individual case. It is a complex disease with limited but growing understanding of multisystem relatedness and disease process connectivity. Much more work and research needs to be undertaken to understand the complex interworking of this disease origin and course (Tinkle et al., 2017).

Please see Table 1 for specific disease processes and the mechanisms of impact proposed for each one.

Those with EDS often identify with the image of a zebra. Many are young and have multiple health problems without an explainable diagnosis. Like Zebras, those with EDS each have a unique presentation to their disease. Most healthcare providers are taught to look for the expected and not unexpected and can often miss this disease in the process (The Ehlers-Danlos Society, 2019).

Table 1	Signs & Symptoms	Pathophysiology	Nursing Care Implications
Musculoskeletal	Pain, muscle weakness, myalgia, fatigue, limited mobility, spinal abnormalities and osteoarthritis (Tinkle et al., 2017)	Symptomatic joint hypermobility, weak joint connections lead to trauma and inflammation to joint articulations (Tinkle et al., 2017)	Early recognition and diagnosis in children can have positive impact on interventions and disease impact over the lifetime (Scheper, Nicholson, Adams, Tofts, & Pacey, 2017)
Pain	Acute and chronic, nociceptive muscle pain, spasms of tendon, generalized pain, dislocations, joint pain, loss of proprioception, GI,, GU, GYN pain, dystonia (Chopra et al., 2017)	See body system subtypes on this table for specific system pathologies	Physiotherapy, low dose tricyclics, stress management, hormonal regulation, topical lidocaine, TENS, opioids are discouraged (Chopra et al., 2017).
Skin and Connective Tissue	All characteristics present to a much lesser degree than other EDS subtypes and include: soft, silky velvety skin, semi-transparent, visible venous structure, hyperextensible and fragile skin (Tinkle et al., 2017)	Disorder of connective tissue matrix proteins, especially collagen with likely collagen genetic malformation (Tinkle et al., 2017)	Surgical wound healing may be delayed (Tinkle et al., 2017)
Gastrointestinal	functional gastrointestinal disorders (dyspepsia, GERD, abdominal pain, diarrhea/constipation). Hiatal hernia, irritable bowel syndrome (Beckers et al., 2016)	Limited studies of underlying cause, some evidence of esophageal dysmotility, visceral hypersensitivity (Beckers et al., 2016)	Assess and maintain optimal nutrition practices
Cardiovascular	cardiovascular autonomic dysfunction: Postural tachycardia syndrome (POTS), neutrally mediated hypotension (NMH) orthostatic hypotension (OH) and orthostatic intolerance (OI) (Hakim et al., 2017)	Low resting blood pressure, limitations and absence of clinical trials, case control studies and population cohort studies for the hEDS subpopulation (Hakim et al., 2017)	Water and electrical replacement, beta adrenergic blockers, postural awareness (Hakim et al., 2017)
Nervous System & Spine	Migraine, Chiari I Malformation, Atlantoaxial instability, Craniocervical instability, Segmental Kyphosis and instability, Tethered Cord Syndrome, Dystonia, Neuromuscular disorders, Tarlov Cyst Syndrome, acquired scoliosis, peripheral neuropathy, Idiopathic intracranial hypertension (Henderson et al., 2017)	lax ligamental structure and poor posture decrease spinal stability, instability and ligament dysfunction at the atlantoaxial and atlantooccipital joints, peripheral nerve connective tissue degradation, reduced intradermal nerve fiber density, tenacin-x deficiency, excess CSF production and reduced absorption (Henderson et al., 2017)	Neck bracing, physical therapy, lumbar puncture, Craniocervical fusion, VP shunts (Henderson et al., 2017)
Gynecological	Menorrhagia, genital mucosal dysfunction, painful intercourse (Tinkle et al., 2017)	Pathophysiology poorly understood (Tinkle et al., 2017)	hormonal therapy as needed, physiotherapy
Dental	TMJ/TMD, limited maximal mouth opening, mucosal bleeding, temporal headache, bruxism (Mitakides & Tinkle, 2017)	Altered teeth morphology, high cusps and deep fissures, enamel hypoplasia (Mitakides & Tinkle, 2017)	Often less response to local anesthetic, key to understand structures to target pain management technique (Mitakides & Tinkle, 2017)
Psychiatric	Depression, anxiety, affective disorder, hopelessness, desperation (Tinkle et al., 2017)	Negative psych state can reduce self-care and exacerbate pain symptoms further (Tinkle et al., 2017)	Validate patient's symptoms, encourage psychiatric counseling, cognitive behavior therapy (Tinkle et al, 20170
Pelvic & Urinary System	Urinary incontinence, pelvic organ prolapses (Tinkle et al., 2017)	Laxity of connective tissue in pelvic floor (Tinkle et al., 2017)	kegels/physical therapy, weight management (Tinkle et al., 2017)
Immune System	Mast Cell activation disorders (Seneviratne, Maitland, & Afrin, 2017)	Mast cells in connective tissue hyperactivity impactive 2+ organ systems, MCAD connection to hEDS poorly understood but clinically significant (Seneviratne, Maitland, & Afrin, 2017)	Encourage exercise in spite of fatigue, Medication management and avoidance of triggers (Seneviratne, Maitland, & Afrin, 2017)

Conclusions

Physical deconditioning can contribute to cycles of weakness and pain exacerbations and disability- promote physical conditioning (Tinkle et al., 2017, p52)

More work needs to be done in the areas of research and scholarship

Primary care should focus on appropriate diagnosis using the updated diagnostic criteria and treatment should be directed at a multisystem, multidisciplinary approach (Caastori et al., 2017)

Management of symptoms varies to differentiate between acute vs emergent symptoms, prevention of complications and management of pain (Tinkle et al., 2017).

References

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