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Diagnosis Delays in Hypermobile Ehlers-Danlos Syndrome and Hypermobility Spectrum Disorders: A Synthesis of Qualitative Literature

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Background

Hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders (hEDS/HSD) represent a group of connective tissue disorders characterized by joint instability, tissue fragility, chronic pain, and multisystem dysfunction leading to disability and reduced quality of life [1,2]. Diagnosis delays of hEDS/HSD are common, and may be 20 years or more [3]. The reasons for these lengthy diagnosis delays is unclear.

Objective

This review sought to identify possible reasons for diagnosis delays by synthesizing the existing qualitative research on hEDS/HSD.

Methods

- Search strategy: PubMed, Scopus, CINAHL, Google Scholar, and Dissertations and Theses databases for all qualitative studies about hEDS/HSD that mentioned the diagnosis process
- Analysis: Sandelowski & Barroso's (2017) process of qualitative research synthesis [4]; primary codes based on the four-factor framework for diagnosis delay by Lapane et al. [5]

Characteristics of Studies

- 283 studies were retrieved from the initial search; 13 studies were included in the analysis.
- Studies were conducted in the UK, Belgium, Norway, Spain, and the US
- Six studies were from the field of rehabilitation, other fields represented included nursing, genetics, psychology, rheumatology, podiatry, community medicine, neuroscience, and child health.
- Six studies showed a diagnosis-related theme or subtheme; in the remaining studies, the diagnosis was discussed within the context of other themes or subthemes.

Findings

Themes related to diagnosis delays were synthesized under four factors:

Disease Factors

- ***Nature of the symptoms***: symptoms are multisystemic, heterogeneous, unpredictable, and mimic other more common conditions
- ***There is no confirmatory test***: the illness is "invisible" to others; with no biomarker, hEDS/HSD is difficult to diagnose with certainty

Patient Factors

- ***Psychological and emotional responses***: chronic undiagnosed, unrelieved symptoms can lead to anxiety, anger, guilt, and depression
- ***Multiple providers - multiple diagnoses***: multisystemic manifestations of hEDS/HSD can lead to each individual symptom receiving a specialty-specific diagnostic label, obscuring the overall picture of hEDS/HSD and delaying diagnosis

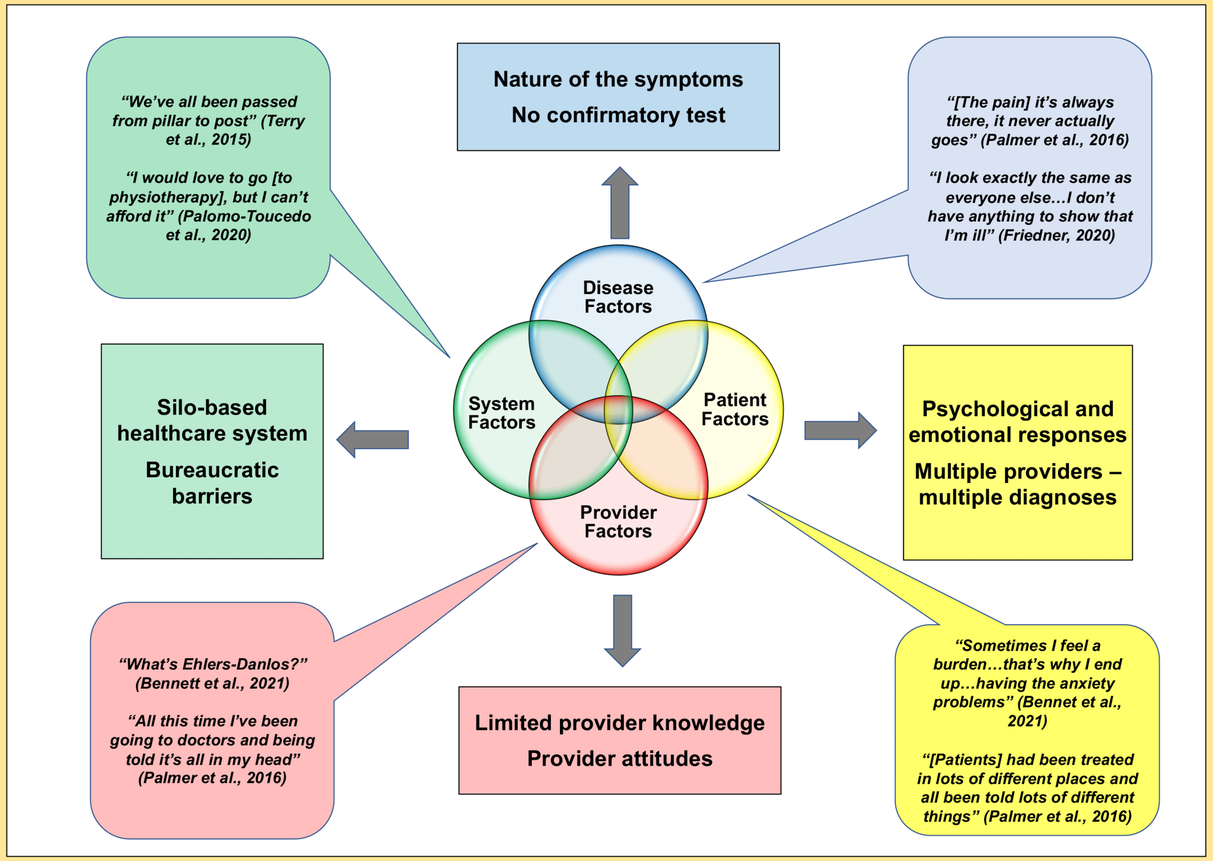
Provider Factors

- ***Limited provider knowledge***: research has evolved rapidly in the past decade; providers may have limited, outdated, or incorrect understanding of hEDS/HSD
- ***Provider attitudes***: participants report lack of empathy and insensitivity from providers; patients may feel dismissed or disbelieved

System Factors

- ***Silo-based healthcare system***: the organization of care remains separated according to clinical discipline, body system, or function; communication between specialties is poor
- ***Bureaucratic barriers***: healthcare access may be limited by cost, time, and long waits for specialty appointments; strict adherence to clinical protocols leave little flexibility for holistic or creative patient-centered approaches

Figure 1. Model of Diagnosis Delays in hEDS/HSD



Conclusion

- hEDS/HSD is a complex, chronic heritable disorder with multisystemic manifestations.
- Multiple factors overlap and interact in ways that contribute to hEDS/HSD diagnostic delays.
- Multidisciplinary collaboration and patient-centered approaches are needed to address the health concerns of hEDS/HSD patients.
- Provider education is critical to prevent diagnostic delays in hEDS/HSD.
- Further research is needed to address:
  - How diagnosis delays impact health outcomes
  - How diagnosis delays impact the patient-provider relationship
  - How diagnosis delays can be reduced in patients with hEDS/HSD

Select References

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SCAN QR CODE for Table of Included Studies