Provider Knowledge, Genetics Referral Patterns, and Diagnostic Delay in Hypermobile Ehlers-Danlos Syndrome

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Introduction

Hypermobile Ehlers-Danlos syndrome (hEDS) is a heritable connective tissue disorder associated with prolonged diagnostic delays. Provider knowledge and genetics referral practices may influence these delays, yet few studies have examined these factors from the patient perspective.

Methodology

Survey Population

- 117 U.S. Adults with hEDS

Data Analysis

- Cumulative link mixed models
- Chi-square tests
- Correlation

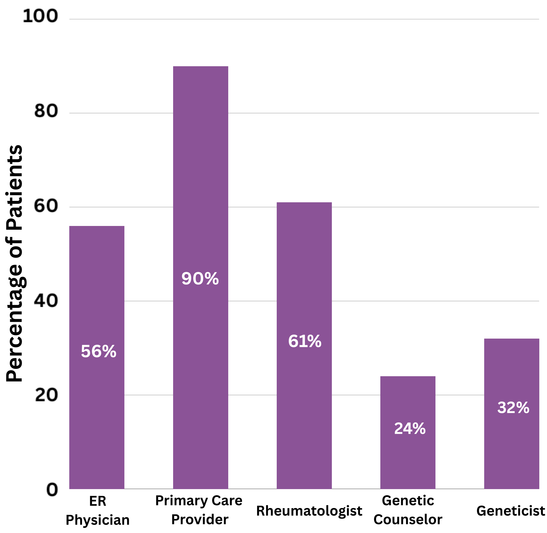
Data Collection

- Online survey assessing provider knowledge, genetics referrals and diagnostic delay

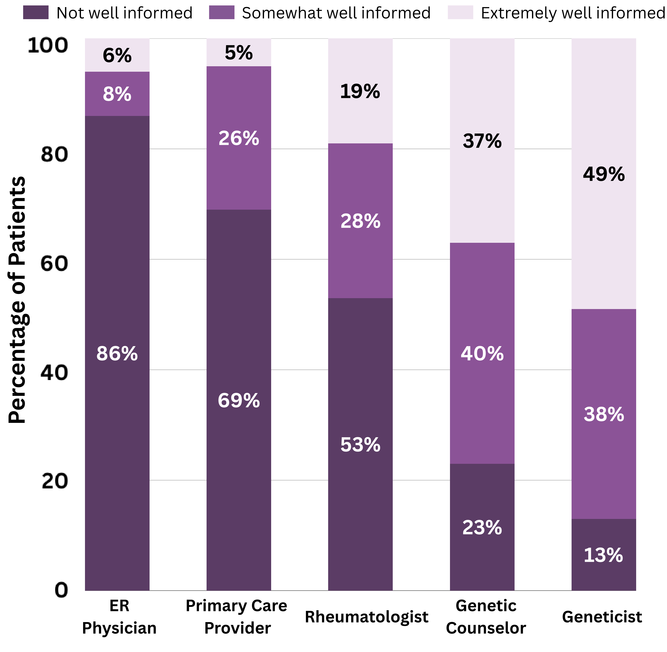
Results

- Participants experienced an average diagnostic delay of 20.7 years (SD = 14)
- Emergency physicians received the lowest patient ratings for hEDS knowledge
- Primary care physicians were the most common source of genetics referrals (28%)
- 27% of participants were never referred to genetics

Providers Seen Before hEDS Diagnosis



Patient Ratings of Provider Knowledge of hEDS



Conclusion

These findings suggest that variability in provider knowledge and genetics referral practices may contribute to prolonged diagnostic delays in hEDS and highlight opportunities to improve provider recognition, genetics referrals, and diagnostic pathways.