Provider Knowledge and Experience in Care, Management, and Education of Pediatric Ehlers-Danlos syndrome

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Introduction

Ehlers-Danlos Syndrome (EDS) represents a family of heritable connective tissue disorders with overlapping phenotypic features, frequently including joint hypermobility, tissue fragility, and skin hyperextensibility.[1,2] Approximately 1 in 2500 to 5000 babies are born with EDS worldwide annually, though this may be a conservative estimate due to the intensive, varied diagnostic criteria for EDS,[3] and a lack of provider familiarity with EDS as a medical disease. Though these defining features are thought to be principal to the identification and diagnosis of EDS, there are 13 recognized distinct clinical subtypes,[1] each defined by both major and minor criteria.

Diagnostically, these EDS subtypes are heterogeneous, both genetically and phenotypically. Definitive diagnosis relies on molecular confirmation with all subtypes except for hypermobile EDS, which is a clinical diagnosis,[1] though a genetic basis is also suspected for hypermobile EDS.[4] Clinical sequalae for patients with EDS include multiple body systems marked by neurologic[5,6], cardiovascular[7], gastrointestinal[8], dermatologic[9], gynecologic[5], and musculoskeletal issues[10]. Additionally, EDS, particularly the hypermobile subtype, is also associated with other co-morbid symptoms, such as chronic pain,[11] deficits with proprioception,[12] headaches,[13] anxiety, and depression.[13]

Diagnostic Difficulties and Delays

Due in part to such complex and multisystem involvement, diagnosis of EDS may be delayed or easily missed by general practitioners and specialists, and there is evidence to show that many are misdiagnosed with other diseases such as chronic fatigue syndrome.[14] Inadequate education and awareness of EDS may contribute to a delay in diagnosis, and/or referral for broad evaluation. In the absence of diagnosis and referral for appropriate care, symptoms of fatigue worsen, and physical deconditioning and mental health can deteriorate quality of life (QoL), increasing the need for more aggressive and costly rehabilitation therapy.[14] As a result of such an interwoven set of diagnostic criteria and associated clinical concerns, children with suspected EDS are most likely to be referred to and seen by a pediatric subspecialist such as a rheumatologist, geneticist, or cardiologist,[15] as opposed to primary management from a primary care or family medicine physician within their community. However, due to a limited number of subspecialists nationally with familiarity of EDS, the referral process results in long delays for diagnosis while poor control of symptoms continues to worsen quality of life.

There have been some recent efforts to inform medical providers about EDS to help aid in symptom management,[16] however, it is currently unknown if general practitioners are comfortable and knowledgeable about EDS and what barriers may prevent optimal care for patients with EDS. The objective of this study was to characterize and identify barriers to practitioner awareness, comfort with care, management, and education of children with EDS.

Methods and Materials

Using REDCap electronic data capture tools hosted at Children's Mercy Hospitals, an electronic survey of 28 questions assessing awareness and diagnostic evaluation of EDS, comfort with care, management, and education

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Table 1. Respondent Characteristics.

Characteristics	n (%)
Clinic role	
Specialty physician	12 (11)
Pediatrician	26 (24)
Family medicine	5 (5)
Pediatric Resident	55 (51)
Nurse practitioner	7 (7)
Other	2 (2)
Clinic location	
Urban	102 (95)
Suburban	5 (5)
Clinic setting	
Academic	106 (99)
Private practice	1 (1)
Experience (by years of practice)	
0-5 years	65 (61)
5-9 years	18 (17)
10 + years	23 (22)
Experience (by # of EDS* cases)	
0	70 (65)
1-3	22 (21)
4-6	9 (8)
7-10	3 (3)
>10	3 (3)
Experience (Currently have patient with EDS*)	
Yes	39 (37)
No	41 (38)
Unsure	27 (25)
Clinical time	
0%-25%	5 (5)
26%-50%	14 (13)
51%-75%	12 (11)
>75%	76 (71)

*Ehlers Danlos syndrome.

of EDS, and barriers that prevented comfort with care, management, and education of EDS was developed along with demographic questions that assessed clinical role and experience. Survey questions used branching logic, gave multiple choices that ranged from "not at all comfortable" to "completely comfortable," and "choose all that apply," while many questions had an "other" category for the respondent to fill-in responses that may not be listed. The survey was electronically sent to pediatric and family medicine practitioners and trainees at 2 tertiary care academic medical centers in one Midwest region of the United States. Participants were asked to respond according to their personal experience, not that of institution, group practices or based on medical literature. Respondents were asked to quantify their experience by years of practice and number of EDS cases managed. The survey was sent on 2 separate occasions over a 2-month period.

The results were analyzed, binary and categorical variables were summarized by frequency and percentage, while the relationship between variables between resident respondents and nonresident respondents were evaluated with T-test. All statistical analysis was completed using SPSS statistics 24 software.

Ethical Approval and Informed Consent

This work was conducted in accordance with the Declaration of Helsinki. Institutional review board approval was obtained from Children's Mercy Kansas City (IRB Study ID: 16060435). All respondents consented when they voluntarily completed the survey.

Results

Respondent Characteristics: Of 107 survey responses analyzed, the respondents were mostly pediatric residents (51%) followed by pediatricians (24%) and specialty physicians (11%). Most the respondents (61%) had less than 5 years of experience while 22% had 10 or more years of experience, and most of the respondents (38%) reported no current patients with EDS that they care for, however, 25% of all respondents were unsure if they currently had a patient with EDS (Table 1).

Familiarity with EDS: Of the surveys analyzed all (100%) reported familiarity with EDS (ie, "heard of" or "learn about"), and respondents most commonly "heard" about EDS from medical text (94%) and from patients (51%). Providers also "learned about" EDS through medical training (79%), from a patient (31%), and EDS seminar/conference (13%).

Caring for EDS and Barriers to Providing Care: Only 9% of all survey respondents were completely or very comfortable with the 2017 EDS criteria, while 4% were completely or very comfortable diagnosing EDS. However, 39% were completely or very comfortable executing plans developed by a specialist, while 9% were completely or very comfortable developing their own plans of care for EDS (Table 2).

Family Education: Of the survey respondents, 29% reported educating families about EDS, but only 7% were completely or very comfortable doing so. The most common barriers that prevent comfort caring, managing, and educating patients with EDS was lack of educational materials, knowledge, and confidence. Fill-in responses indicate that most respondents would like education (in the form of informal and formal didactics and reading materials) to overcome the identified barriers (Table 2).

Table 2. Ehlers Danlos syndrome (EDS) Survey Question Responses.

Respondent responses	Yes n (%)	Not comfortable n (%)	Somewhat comfortable n (%)	Very comfortable n (%)
Hearing about EDS				
Have you heard of EDS	107 (100)			
Did you hear about EDS from Medical text	101 (94)			
Did you hear about EDS from Patient	55 (51)			
Did you hear about EDS from social media	9 (8)			
Learning about EDS				
Did you learn about EDS from medical text	107 (100)			
Did you learn about EDS from Patient	33 (31)			
Did you learn about EDS from medical training	84 (79)			
Do you currently have patients with EDS	39 (36)			
Caring for EDS				
Understanding of 2017 EDS criteria		62 (58)	35 (33)	9 (9)
Diagnosing Patients with EDS		68 (63)	35 (33)	4 (4)
Executing Plans of Care a Specialist develops		20 (19)	45 (42)	42 (39)
Making Plans of Care for EDS		62 (58)	36 (34)	9 (8)
Barriers that prevent comfort in caring for EDS				
Lack of educational materials	44 (41)			
Lack of knowledge	51 (48)			
Lack of confidence	57 (53)			
Lack of time	26 (24)			
Barriers that prevent comfort in managing EDS				
Lack of educational materials	37 (35)			
Lack of knowledge	62 (58)			
Lack of confidence	71 (66)			
Lack of time	34 (32)			
Education about EDS				
Do you educate families about EDS	31 (29)			
Educating families about EDS		60 (66)	28 (26)	7 (7)
Barriers that prevent providing education about EDS				
Lack of educational materials	50 (47)			
Lack of knowledge	71 (66)			
Lack of confidence	61 (57)			
Lack of time	38 (36)			

Not comfortable includes "not at all and not very" categories. Very comfortable includes "very and completely" categories.

Residents versus Non-Residents: Since residents made up half of the survey respondents, the respondents were split between residents and nonresidents and compared. There were significant differences noted in clinical time ($P < .001$), with residents reporting more clinical time, and years since training completed ($P < .001$) with residents reporting no time since training completed. Residents reported more comfort executing plans developed by a specialist ($P < .048$) and less comfort making their own care plans for patients with EDS ($P = .001$). Additionally, residents were less comfortable educating families with EDS ($P = .001$) and had less interest in attending educational workshops or webinars about EDS ($P = .033$).

Discussion

To our knowledge, this is the first survey of medical providers that assesses knowledge, comfort with care, management, and education of patients with EDS, and barriers to care, management, and education for patients with EDS. Surveyed providers demonstrated a general familiarity with EDS that was obtained from medical texts and medical training, which is encouraging given the perceived rarity of the disorder; however, most providers expressed lack of knowledge and confidence as a barrier to care, management, and education for those with EDS. Residents reported more limitations due to lack of knowledge and confidence as a barrier to care, while nonresidents reported more time constraints and

lack of knowledge as main barriers to care and education. Most respondents in our sample reported no current patients with EDS, however, many were also unsure if they were caring for a patient with EDS. Among all respondents, including those not currently caring for a patient with EDS, many report feeling very comfortable executing plans from a specialist as opposed to developing their own plans. There were also differences seen in plans of care for patients with EDS as residents were less comfortable executing plans developed by a specialist and making their own plans of care compared to nonresident respondents, which likely represents practitioner experience.

It is unknown which providers, if any, have primary responsibility for patients with EDS as patients with EDS typically consult with multiple subspecialists, and the ownership of coordination likely falls on the primary practitioners by default; alternatively, in the absence of medical ownership, patients and parents may be forced to coordinate their care and potentially conflicting treatment recommendations. Primary practitioners are well positioned to help patients with EDS navigate the assessment process and initiate intervention, while also continuing to provide ongoing management and care. However, there are several barriers to address to promote increased EDS management by a community or primary care physician. First, many providers were unsure if they were caring for a patient with EDS, which may reflect a lack of foundational knowledge to recognize EDS. Second, providers reported difficulty with identified educational materials to inform EDS care. Third, residents were found to have less comfort educating families about EDS compared to nonresident respondents, which is likely due to practitioner experience. Interestingly though, residents reported less interest in a local workshop or webinar about EDS, which likely reflects a current educational situation where they focus on basic required knowledge prior to building knowledge in rare diseases. This could also reflect a mischaracterization of EDS as a "rare disease," despite the fact that some forms of EDS, particularly the hypermobility subtype, are likely less rare than previously thought,[3] and thus a need to better integrate it into general medical education curricula. The knowledge barriers that exist, low number of respondents who have learned from an EDS conference, and nonresident interest in a workshop or webinar shows a prime opportunity to development and implement an EDS educational program to meet the practitioners needs.

While subspecialty care and multidisciplinary care teams[17] are integral to the diagnosis, care, and management of patients with EDS, there is a crucial role for primary practitioners as well. A diagnostic and management process that over relies on specialist care is wrought with limitations that affect patient access and care, including delayed access due to lengthy waitlists, delayed initiation of appropriate treatment, and family frustrations in attempting to integrate recommendations from multiple separate providers. Though respondents were much more comfortable executing plans developed by a specialist rather than developing their own care plans for patients with EDS, the availability and sustainability of multidisciplinary care is multifactorial,[18] which may lead to issues with long-term, and immediate or acute patient needs. This represents an opportunity to develop educational resources to bolster primary practitioner knowledge and confidence in the care and management of patients with EDS. Barriers identified in this study, such as inadequate education and lack of confidence with EDS, may contribute to a delay in diagnosis, and referral for unnecessary evaluation, which may lead to negative outcomes and poor quality of life for patients with EDS.[19] To address these barriers, targeted educational materials and on-going models for consultation and supervision could be developed and implemented to improve comfort with care, management, and education of EDS within a primary care, family medicine, or community practice, which could lead to improved care and better outcomes.

Our study has several limitations, which includes that the survey was completed locally, and the findings may not be generalizable to larger groups of practitioners in different geographical regions. About half the respondents were residents, which represents an early stage in medical training and may bias some of the results, however, we did separate the responses between residents and nonresidents and the findings were very similar. Additionally, our institution has a multidisciplinary EDS clinic that serves the region which could bias the results in 2 ways: (1) increased awareness of EDS by the respondents; and (2) a higher degree of reliance on specialty care than regions without a multidisciplinary EDS clinic. In general, we expect the barriers reported in this study to be present and potentially more profound in areas without a multidisciplinary EDS clinic.

This is the first study to evaluate providers awareness, understanding of care, management, and education of children with EDS. All respondents have heard and learned about EDS, but most are not comfortable with diagnosis, care, management, and education of children with EDS. Barriers to care and management include lack of educational materials, knowledge, and confidence, which could potentially be improved through educational materials and programs that are EDS specific. More research is needed to confirm these findings and determine optimal educational modalities and implementation.

Author Contributions

JJ and WB equally contributed to the conception, drafting and final version of the whole manuscript. All authors read and approved the final manuscript.

Declaration of Conflicting Interests

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