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ARTICLE

An evaluation of practices and policies used in genetics clinics across the United States to manage referrals for Ehlers-Danlos and hypermobility syndromes

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ABSTRACT

Purpose: Hypermobile Ehlers-Danlos syndrome (hEDS) and joint hypermobility syndrome (JHS), among other conditions, comprise a collection of heritable disorders of connective tissue. There are recognized challenges in diagnosing JHS/hEDS. Despite a lack of identifiable molecular etiology for these conditions, referrals to medical geneticists for evaluation are commonplace, and they continue to rise. Because of an absence of nationally recognized referral guidelines for JHS/hEDS, health care institutions are left to develop their own policies. The purpose of our study was to characterize these ad hoc policies systematically and at a nationwide level.

Methods: We conducted a mixed-methods study of 71 board-eligible or board-certified genetic counselors, including 15 qualitative interviews.

Results: Cross-case analysis revealed multiple motivations for creating these policies, methods to more effectively manage referrals for hypermobility and concern for EDS, and participants' evaluations of the successes and shortcomings of these policies at their institutions. We found diverse and unstandardized policies that were meant to address numerous perceived challenges. This lack of standardization is a concern because it may result in inconsistent access to care for patients with JHS/hEDS and create barriers to diagnosis and treatment.

Conclusion: Our findings demonstrate that policies vary widely, and genetic counselors are concerned about the potential impact of this variability on the quality of care for patients with JHS/hEDS.

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Introduction

Among the many heritable disorders of connective tissue, the most common may be hypermobile Ehlers-Danlos syndrome (hEDS). This disorder can be characterized by joint hypermobility, stretchy or fragile skin, and general weakening of tissues such as joints.[1] Overlapping symptoms, multisystemic involvement, and extensive variation in clinical phenotypes present diagnostic challenges for even well-trained professionals,[2] suggesting that referring patients to a specialist may be the most efficient way forward.[3] The symptoms that can accompany multisystemic diseases, which are sometimes nonspecific in nature, can lead to referral indications that are broad and ill defined. Additionally, hEDS lacks a known molecular etiology; therefore, no genetic test is available to streamline diagnosis,[4] and clinicians rely on robust but complex clinical diagnostic criteria.[1]

Joint hypermobility syndrome (JHS) is a diagnostic category that is no longer recognized by international scientific standards. There is a significant overlap between the former JHS phenotype and hEDS, and the 2 disorders are now clinically treated as a single entity.[5] However, many clinicians continue to use the label JHS because of the complex and recent nature of the updated diagnostic criteria, frequently referring these patients to medical genetics for further assessment (see below). In this article, we therefore use the combined term JHS/hEDS to refer to this population.

These factors may explain the growing number of EDS referrals to general genetics clinics.[6] Because of the general shortage of genetics professionals,[7] time-intensive nature of hEDS appointments,[8] and high prevalence of symptomatic joint hypermobility in the general population,[9] providing care to these patients can strain available clinical genetics resources. In response to these challenges, some health care systems have radically reduced the number of clinic hours available for EDS referrals and implemented referral triage measures to prioritize higher risk patients.[10] Given the potential impact of these policies on patient care, we developed a mixed-methods study to evaluate both the motivations and the practices that institutions have established in this regard. To our knowledge, our study is the first to address either question. We have found significant variability in policies that worried our participants. These results suggest potential utility for standardization across institutions, and further study is warranted to assess the value of creating nationally recognized JHS/hEDS practice guidelines to assist clinicians in providing the best care for this patient population.

Materials and Methods

Recruitment

We designed an exploratory study using a descriptive qualitative approach. This study was approved by the Virginia Commonwealth University Institutional Review Board. We recruited participants based on convenience and snowball sampling, through the membership directory of the National Society of Genetic Counseling. Candidates included board-eligible or board-certified genetic counselors who work in general genetics clinical settings within the United States. Additional inclusion criteria were that participants spoke English and were 18 or older.

Candidates for qualitative interviews were selected purposefully from a subset of survey respondents to capture a broad institutional and geographic scope of perspectives. There is not a universally accepted sample size for qualitative studies because a significant outcome is often dependent on achieving saturation of themes.[11] According to similarly designed qualitative studies, 12 to 15 interviewees are normally sufficient to reach saturation,[12] and we anticipated needing to recruit accordingly.

Study design

Our survey included multiple-choice questions and free-text prompts to generate preliminary quantitative information regarding relevant policies (see Supplemental Survey). Responses were collected through REDCap, a secure application developed for the management of such online surveys. Participants were incentivized by a drawing for 1 of 3 VISA gift cards, valued at $40, $30, and $20.

One team member (L.B.) contacted a subset of the survey respondents who expressed interest in completing a semi-structured interview, which was conducted through Zoom. Interview participation was incentivized by a guaranteed $20 VISA gift card. The interviews gathered qualitative information regarding the participants' institutional referral policies and knowledge of motivations behind those policies (see Supplemental Survey).

Data analysis

Qualitative interview data were analyzed with inductive reasoning and cross-case analysis.[11] Categories were identified and compared across all participants via a manual review and then analyzed in a multistep process to reduce researcher bias.[11] Two researchers (L.B., B.S.) independently coded all the data and achieved consensus about the themes and categories. Two data auditors (D.G., J.Q.) then reviewed their consensus and made revisions.

Results

Seventy-one individuals completed the survey. They came from at least 59 unique institutions, with 2 participants choosing not to provide this information. Using purposive sampling, we conducted a total of 15 interviews with the survey respondents (63%), each representing a unique institution. Interviews ranged in length from 19 to 59 minutes. Data represented in this section come from both surveys and interviews. Personal identifiers have been removed, but a unique number for each participant is given after quotations.

Four primary categories of results emerged from our data: motivations for developing EDS-related practices and policies, types of policies implemented, their perceived outcomes, and a call for the development of formalized national guidelines (see Table 1 for representative quotes).

Motivations for practices and policies

Fifty-five survey respondents (77%) worked at institutions that currently had policies or practices in place to manage the volume of referrals specifically for JHS/EDS. Thirteen institutions (18%) did not evaluate patients for JHS/EDS without other syndromic or vascular indications. Based on a thematic analysis of interviews, we found that the motivations behind these policies included logistical, personal, and professional issues surrounding the evaluation and management of referrals for this population.

Fourteen interviewees (93%) disclosed that a perceived large volume of JHS/EDS referrals logistically prompted the development of policies. Nine participants (60%) mentioned the lack of available clinicians to perform the evaluations. Others (67%) believed that JHS/EDS appointments for evaluations took longer compared with other, nonurgent indications. They correlated the extended visits with the need for psychosocial support, education about testing options, and the taking of complex histories. These institutions developed policies to ensure that patients with other indications could still be seen in a timely fashion.

Personal reasons were also seen as motivators. For instance, provider preferences were discussed. In some cases, genetics providers only wanted to see patients with conditions of special research or practical interest rather than patients with JHS/EDS. In some institutions, only 1 or 2 medical geneticists were willing to evaluate these patients. In other instances, certain providers specialized in connective tissue disorders; therefore, those individuals took on the responsibility of seeing all such referrals, limiting the number of available appointments. Provider dissatisfaction (40%) was another personal reason motivating the development of JHS/EDS-related policies, with participants citing burnout and frustration working with this population.

Professional motivations were also present, and participants disagreed about whether and how genetics departments might provide care for patients with JHS/EDS, which affected policy development. Twelve interviewees (80%) referenced the current lack of a known molecular basis for hEDS specifically as making them feel they were unable to affect care for these patients significantly. The lack of subsequent treatment options was believed to be frustrating for patients as well.

On the other hand, some providers felt that they could and do have a significant impact on this patient population's care and did not perceive the lack of genetic testing for JHS and hEDS as a source of patient dissatisfaction. They highlighted other ways in which genetics providers can provide important services for the EDS population, such as through education and emotional support.

Finally, most institutions (*N* = 12, or 80%) reportedly implemented their policies as a way to improve the quality of patient care. This was in large part (67%) accomplished by managing patient expectations and combatting "unrealistic expectations" (31). For instance, one participant explained that in their practice, they begin discussions with patients being evaluated for hEDS by explaining "a lot of times, genetic testing is not ordered because the diagnosis can be made without genetics or without a blood test. And that has, I think, reduced some patient dissatisfaction" (12). Another way these policies sought to improve patient care was by educating clinicians about appropriate referrals and by requiring patients to complete previsit questionnaires.

Practices and policies

Scheduling policies

Ten participants (67%) reported that it was their institution's policy to limit how many patients with JHS/EDS they evaluated within a given period. Some clinicians also strategically scheduled these appointments at specific times during their clinics to allow for the extra time required to address these patients' needs, for instance by reserving the final appointments of the day. Some institutions even created a separate clinic for evaluating these patients so that there would be no competition for appointments between patients with JHS/hEDS and patients with other indications. Finally, 3 interviewees (20%) reported that their institutions sought to limit time committed to patients with JHS/hEDS by declining to offer this population long-term follow-up care, something they do for other conditions with clinical diagnoses.

Triaging referrals

Many institutions had policies to manage the number and source of referrals for JHS/EDS. The majority rejected self-referrals to "triage some of the volume" (12). Three participants (20%) shared that their institutions created their own set of criteria to determine whether they would evaluate patients with JHS/EDS in their clinics. Thirteen survey respondents (19%) would not evaluate patients for JHS/EDS without other syndromic or vascular indications, although 3 interviewees mentioned that exceptions to this policy were made for individual cases. Similarly, 5 institutions (33%) only accepted referrals for JHS/EDS from certain specialties or providers that they assessed as more knowledgeable about appropriate referrals to genetics.

Seven institutions (47%) sent letters to referring providers which served a variety of purposes, including provider education about JHS/hEDS and how to perform the

Table 1 Representative quotations

Category	Subcategory	Quotation
1 Motivations	1.1 Logistical Motivations	"Even with two providers who were seeing patients a few days a month, we just didn't have the capacity to see everybody who wanted to be seen" (56).
		"It can take an hour and a half, and then […] you're already running late for your next patient and didn't even finish what you need to talk about" (29).
	1.2 Personal Motivations	"We have one provider who sees patients for connective tissue conditions, and he's only in clinic two days a month, so it's a limited capacity for this indication" (56).
		"I mean, to be honest, it's just, it's really […] tiring. Like, it's a hard patient population" (71).
	1.3 Professional Motivations	"They've waited six-plus months to get in the clinic, and then they show up and we don't have any testing to offer them, and then they're not happy about that" (70).
		"Whether they have an EDS diagnosis or not at the end of the appointment, it really doesn't change or fix some things for them. And I think that is probably disappointing or tough for them" (31).
		"I believe in the power of a diagnosis, and I think that that means something. And I think for these patients, I have really a lot of empathy and a lot of compassion for their journey and their struggle, and a lot of them have had really tough experiences, and it's a lot getting to us. And they share […] how much they want an answer and how much they want to understand what is happening to them" (17).
		"They're often pretty grateful for our services, and they tend to say […] 'Thank you for all of this information.' I feel like I've done like nothing, but they tend to say, 'Thank you so much.' And, you know, we make recommendations about physical therapy and stuff like that and they find those types of things helpful" (07).
2 Practices and Policies	2.1 Scheduling Policies	"The clinic is weekly, there's a set amount of appointments each week, and of those appointments, only half of them can be connective tissue referral related" (12).
		"Every provider in our clinic has one slot for [a] clinic session that is designated only for EDS patients. So, for the average provider who has like a half-day clinic session, we're usually seeing like five or six new patients up to that, and one of those would be always EDS" (17).
		"We decided to put them as the last two patients, presumably, of the clinic, so that way they could have the time that they deserved and needed to be seen. […] That way we wouldn't get too far behind with the rest of clinic" (15).
		"We can help diagnose, but we're not going to treat because we don't have the time that we can devote to that really with our clinic" (15).
	2.2 Triaging Referrals	"If we get a referral for someone – and it happens very frequently – where it just says like 'hypermobility' or 'rule out EDS,' like joint laxity, things like that, we have a […] letter that we've created that just explains that hypermobility is a common feature in the general population and we don't have the genes identified for hypermobile EDS, and so because of that there, we don't see people for that indication since there's no testing that we can provide" (49).
		"We review the referral to see if they meet our criteria to even get scheduled […] we have written criteria and if you don't have one of those things, then we don't have anything to offer you for connective tissue evaluation" (56).
	2.3 Pre-Visit Requirements and Patient Questionnaires	"We actually have a triage nurse that […] gets all the paperwork, and we have an online work queue, and we keep track of everybody that has sent in an echo[cardiogram] and an eye exam" (37).
		"The questionnaire is kind of converted into a review of systems summary that an intern makes for us and that is copied directly into the note" (17).
		"The first page of that questionnaire is kind of a red flag checklist where it's a shorter number of questions, like six questions or about, and essentially if the answer is yes to any of those things, then that would indicate that the patient should be moved into our general kind of queue" (17).

(continued)

Table 1 Continued

Category	Subcategory	Quotation
3 Perceived Outcomes of Policies	—	“I think it has protected our regular clinic appointments for patients that we really feel like we need to see, and we have things to offer for them. So, I think that feels like a little bit more equity” (70). “People who used to send tons of patients for hypermobile EDS now don’t because they know we won’t see [them], and those providers also provide better guidance to their patients. […] So that’s really nice because people are less angry about not being able to see us because we’ve done a better job of explaining why” (56). “The downside of not seeing everybody is that there will be some folks that we miss that maybe we should have seen” (56).
4 Calls for Professional Guidelines	—	“We probably do need an organization to put out some more official rules, so that way we’re not just all clambering to come up with our own best policy. It would be better if there was a national recommendation for that” (49).

EDS, Ehlers-Danlos syndromes.

evaluation for a clinical diagnosis. Other letters explained why a genetics evaluation would not be indicated for their patient, for instance, by including the diagnostic criteria for hEDS along with several important academic articles, demonstrating that the diagnosis does not require a referral.

Previsit requirements and patient questionnaires

The majority of participants reported that their institutions officially requested specific evaluations before a consultation, including, for example, a rheumatology evaluation, echocardiogram, and/or dilated eye examinations. Five participants (33%) stated that patients would not be scheduled until these documents were received, whereas others said that although requested, they were not technically required.

Eight respondents (53%) used questionnaires that either were created specifically for the JHS/hEDS patient population or included JHS/hEDS-specific questions. These questionnaires could be quite extensive, with some upward of 20 pages in length, and included questions about symptoms, recent specialist visits and examinations, and family history information.

The majority of participants used the questionnaires to replace the intake during the genetics appointment and thus saved time for more pressing issues. Some institutions also used their questionnaire to create a targeted medical record review so that they could more efficiently access the information during evaluations and assist in triaging patients. Questionnaires also helped to set patient expectations for the appointment with genetics.

Utilization of other staff

Eight interviewees (53%) reported that they utilized additional staff to manage care for the JHS/hEDS patient population. These supplementary staff included nurses, scheduling teams, nurse practitioners, physician assistants, and medical/genetic counseling assistants. They often received specific guidelines or training on how to handle referrals for the JHS/hEDS patient population and thus served to ease workflow by triaging referrals, sending letters to patients or providers, tracking received documents, performing a clinical exam of patients with JHS/hEDS, etc.

Three institutions (20%) had implemented a 2-step appointment for the evaluation of patients with JHS/EDS, with 2 separate patient sessions occurring on different days, to optimize utilization of physicians’ time. Two of those institutions used a genetic counselor for a preliminary visit before scheduling an appointment with a physician. The genetic counselor would only be involved in the second session if genetic testing was recommended. The third institution had a preliminary phone call with the genetic counselor before scheduling a physical evaluation with a medical geneticist. Similarly, 4 participants reported that their institutions had changed their clinic flow so that patients were evaluated by nurse practitioners or physician assistants specifically hired to work with this population.

Perceived outcomes of policies

Overall, most participants reported that limiting the frequency or availability of appointments for patients with JHS/hEDS had the intended effect of reducing provider dissatisfaction and shortening wait times for other referrals. One respondent mentioned that their institution had seen a reduction in the number of referrals from providers to whom they had sent educational letters. Another noted that their institution's policies allowed for both patients and providers to have some of their goals met—reducing wait times and not being overwhelmed with JHS/EDS referrals—"even if neither goal is winning out over the other" (17).

Seven participants (47%) reported that their policies have helped to increase the efficiency of patient sessions, as well as overall clinic flow, citing the questionnaires as valuable in this endeavor. "We just have a little bit more time to chat with the patient," said one interviewee (29), and another noted that they can have "a more productive conversation" (17). Our participants also described how staff utilization changes allowed for more efficient use of genetic counselors' and medical geneticists' time.

Certain negative outcomes of these policies were also identified. Longer wait times for patients with JHS/hEDS were mentioned by 10 interviewees (67%). One participant worried that their policy of requiring prior evaluations before scheduling patients had resulted in substantial delays due to missing information. "In our work queue of referrals, we probably have almost two hundred referrals that are incomplete for all those people" (37).

Several participants (33%) mentioned the possibility of missed diagnoses as a result of their policies. One participant asked, "What if we were missing patients who would then have an aneurysm or rupture? […] Could we have had an impact on preventing an outcome like that?" (17). Similarly, another participant worried that patients may no longer have access to proper screening or long-term management because of their institution's policies (12). Furthermore, some participants voiced their discomfort with having these separate policies. "There are issues any time you're treating a patient referral differently or having a different kind of process. So anyway, I guess [I have] discomfort with our process and feeling like it's not enough, but [it's] also the only way that we can see these patients" (17). The idea of a multidisciplinary clinic was proposed by several participants, which would include genetics as a more efficient and effective way to care for this population.

Calls for professional guidelines

Although institutions reported a wide variety of perceived challenges in caring for patients with JHS/hEDS, 11 (73%) institutions' practices and policies were the product of trial and error and were constantly evolving. Participants noted that the lack of professional guidelines posed a significant difficulty for providers who were doing their best to create their own criteria to minimize missed diagnoses. "Everyone kind of has to come up with their own guidelines," one interviewee complained, noting a need for "some more official rules, so that way we're not just all kind of clambering to come up with our own best policy. I just feel like it would be better if there was a national recommendation" (49).

Discussion

Our study is the first to our knowledge to address the growing incidence of clinics creating policies to streamline referrals for patients with JHS/EDS. Seventy-one board-eligible or board-certified genetic counselors who work in a general genetics setting completed surveys, and 15 completed qualitative interviews on these topics. Using these data, we examined both the motivations behind seeking to create JHS/EDS referral policies and the specific forms those policies took.

Over three-quarters of survey respondents reported having explicit policies or practices in place to manage the volume of JHS/EDS referrals, and 8% declined to evaluate individuals referred for these indications. Even when they did see these patients, one-fifth provided no long-term follow-up care. Policies were overall perceived to have successfully reduced provider dissatisfaction and wait times for other referrals, although concerns about lack of standardization were raised, and these policies indicate that this population of patients is treated differently from the rest.

Our data suggest that the primary reason (reported by 93% of participants) for the implementation of these policies was the perceived large and increasing volume of referrals that genetics clinics receive for these indications. This increase in JHS/EDS referral rates to general genetic clinics has been reported by others who have hypothesized that it could be due to the complexity of the multisystemic condition and the difficulty in diagnosing it.[2,3,6] Additionally, the overall incidence of symptomatic hypermobility may be quite high in the general population, even while it goes underrecognized and undertreated.[13] There is significant symptom overlap between the different subtypes of EDS.[1] This overlap may influence nongenetics providers to proceed with caution and refer to genetics to attempt to rule out other subtypes of EDS, such as vascular EDS, which have a known molecular etiology. The perceived increase in JHS/EDS referrals coupled with the general, widely recognized issue that resources in medical genetics are already insufficient overall[7,10] makes the issue of personnel and workflow management particularly pressing. Moreover, these paired concerns suggest that limitations on genetics practice will become even more acute in the future as demand from JHS/EDS and other patient populations continues to grow.

In addition to seeking to manage workflow, some clinics adopted referral policies because only a small subset of their clinicians was willing and able to evaluate patients with JHS/hEDS. This was because of both limited expertise and personal preferences. The majority (67%) perceived the care they were able to provide to this population to be unimpactful, especially because of the lack of molecular genetic testing for

JHS and hEDS. Additionally, some providers felt that genetics would not be able to offer much in terms of treatment or medical management. These limitations frustrated patients, and for clinicians, they led to feelings of burnout, which is known to worsen quality of care, increase medical errors, and decrease patient and provider satisfaction.[14] Although it is not possible to ascertain the source of patients' dissatisfaction from our data, it is possible that they consider genetics responsible for the multidisciplinary care they are seeking, as such a desire in this population has been well documented by other studies.[15,16] The question of the most appropriate medical home for these patients has been raised.[17] However, JHS/hEDS care likely cannot be managed by one department alone, such as genetics, because of these patients' complex and lifelong medical needs. This study demonstrates the importance of communication across multiple specialty care providers in supporting the needs of the JHS/hEDS population. Thus, a multidisciplinary clinic rather than a sole department would likely be of greatest benefit to this population.[16,18] It is important to note that these feelings were not universally shared. In contrast, some providers believed that they made a significant impact on this population's care by providing a clinical diagnosis, psychosocial counseling, and medical management recommendations. A recent study has similarly found that some genetic counselors feel caring for patients with JHS/hEDS is particularly rewarding because of the psychosocial dynamics involved.[8]

Institutions undertook a variety of policies to manage JHS/EDS referrals to their clinics. The majority (67%) limited how many appointments they reserved for evaluations of patients in this population. Although this statistic is novel, other studies have reported similar cases, such as the fact that specific clinics limit 1 day per month to evaluate potential adult EDS patients.[10] Some even created separate JHS/hEDS clinics to ensure that other patients could still be seen in a timely fashion.

Most of our participants (67%) believed that clinic visits evaluating patients for EDS often took significantly more time than other indications of similar urgency, which is supported by other research.[10] To accommodate this, some scheduled these patients at specific hours to ensure that there would be enough time for a thorough and appropriate assessment.

To address inappropriate referrals, approximately half of our participants (47%) sent JHS/hEDS-specific letters to referring providers. These letters were designed to educate providers about the condition, clinical criteria to diagnose, medical management, and specific institutions' criteria for appropriate referrals. Participants believed these policies had been successful and improved the quality of patient care indirectly by educating their existing care team. The desire to educate providers has been endorsed by Eckstein et al,[8] who found that genetic counselors in their study called for more education and training on hEDS for all health specialties.

In addition to requiring specific medical evaluations (eg, rheumatologic and dilated eye examinations or echocardiograms) before scheduling a visit, the slight majority (53%) of our participants sent questionnaires to patients beforehand to verify the appropriateness of their referrals and to streamline consultations. These questionnaires were often extensive, asking patients to detail symptoms, recent specialist visits, and family histories. Questionnaires were also designed to set patient expectations by ensuring that both parties' goals were aligned, and transparency was provided surrounding potential outcomes of the visit. These policies were also seen to improve efficiency, productivity, and patient care because they allowed for more time during the clinic visit to address education and counseling, which were perceived to be the most valuable part of the appointment for this population.

Some institutions also hired additional staff to perform clinical evaluations for these referrals instead of a medical geneticist. The inclusion of providers, such as an advanced nurse practitioner, was also seen as successful, and it has been shown to improve access, satisfaction, and chronic disease self-management.[19] Some institutions implemented a 2-step process, in which patients first met with additional staff before seeing a physician. Such policies prevented or reduced the burden on medical geneticists' time, allowing them to operate at the top of their scope of practice without creating barriers to the patient population's care.

However, participants acknowledged certain shortcomings and negative outcomes of JHS/EDS-specific referral policies. Missed or delayed diagnoses were participants' primary concern for having any type of restrictive or exclusionary criteria. Other worries included longer wait times and increased patient dissatisfaction. These concerns are supported by evidence from a wide array of research looking at the lived experience of patients with EDS.[20-25] Care for this population presents unique challenges for a number of reasons, including diagnostic criteria, psychosocial dynamics, and workflow. Our research underscores that all medical professionals, including genetics providers, must work to improve how they support this patient population, whether it be directly or indirectly.

Although some participants mentioned that they consulted the 2017 diagnostic criteria,[1] institutional policies were often developed ad hoc by medical geneticists based on past clinical experience in lieu of nationally recognized referral guidelines. In fact, nearly three-quarters (73%) of the policies were developed through trial and error and were liable to change. Participants experienced distress and worried that these diverse and potentially conflicting policies could result in inconsistent standards of care for patients with JHS/EDS and create additional barriers to already scarce and often inaccessible care.[26] These results suggest potential utility in the standardization of these practices across institutions.

Limitations

Our study is exploratory, and we have found a vast diversity and inconsistency of policies in place across institutions. These data are not necessarily generalizable to all institutions across the United States, given the scope of the study. Additionally, some participants were not directly involved with the creation of the policies at their institution; therefore, they could only report based on their current knowledge and awareness of these matters, which may be incomplete.

Conclusion

Our study has showcased the perceived challenges in managing care for this patient population. We found a diverse and unstandardized array of practices and policies that institutions have used to manage these referrals. Although genetics providers are working to educate patients and clinicians, as well as screen and triage patients efficiently, they report limitations in the form of insufficient resources in both time and personnel. There is also a lack of nationally recognized JHS/hEDS referral guidelines. These limitations are both clinically and ethically concerning because they may result in an inconsistent and unjust standard of care for patients with JHS/hEDS, create additional barriers to clinical support, and result in missed diagnoses. Our data provide a better understanding of the perceived challenges genetics institutions and health care providers are facing and the practices and policies they have undertaken to improve those issues from their individual standpoints. They offer a foundation for future quantitative research to support the creation of nationally recognized guidelines, which would ultimately have the potential to enhance and standardize the care for the JHS/hEDS patient population across the country. Nationally recognized guidelines have the potential to increase nongenetics providers' awareness of when a referral to genetics is appropriate, which is critical to reducing the likelihood of missed diagnoses for molecularly defined types of EDS. Furthermore, by decreasing inappropriate referrals, guidelines may also streamline and reduce the time to diagnosis for individuals with JHS/hEDS.

Data Availability

The corresponding author will supply our data individually upon appropriate request.

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Author Contributions

Conceptualization: L.B., B.N., D.G., J.Q.; Methodology: L.B., B.N., D.G., J.Q.; Investigation: L.B.; Analysis: L.B., B.N., D.G., J.Q., D.D., C.M.E.H.; Writing: L.B., B.N., D.G., J.Q., D.D., C.M.E.H.

Ethics Declaration

This study was approved by the Virginia Commonwealth University Institutional Review Board and adhered to the principles of the Declaration of Helsinki. Informed consent was obtained from all participants as required by the IRB.

Conflict of Interest

The authors declare no conflicts of interest.

Additional Information

The online version of this article (https://doi.org/10.1016/j.gimo.2024.101960) contains supplemental material, which is available to authorized users.

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