Access to Genomic Medicine: Perspectives from the Ehlers-Danlos Syndromes and Hypermobility Spectrum Disorders Support Community

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The **Ehlers-Danlos** Society.

The Ehlers-Danlos Society is a non-profit organization dedicated to saving and improving the lives of those affected by Ehlers-Danlos Syndromes (EDS), hypermobility spectrum disorders (HSD), and related conditions.

Background

Genomic medicine enables patients' genetic information to inform clinical care, such as testing, treatment, and counseling, yet many patients and caregivers face barriers to access. Even among healthcare providers who incorporate genomic medicine into their practice, patients and caregivers encounter obstacles (Obeng et al., 2018; NHGRI, 2019). Identifying and addressing barriers among those with rare diseases is the aim of Inspire's collaboration with BCH on a NIH-funded research project.

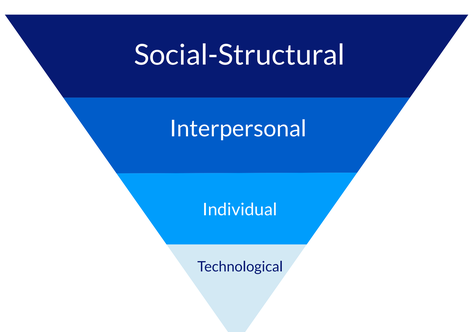
Challenges accessing genomic medicine increase with rarer conditions, such as Ehlers-Danlos Syndromes (EDS), a group of hereditary disorders of connective tissue that vary in the ways they affect the body and in their genetic causes. Patients and caregivers discuss these challenges in the Inspire Ehlers-Danlos Syndromes and Hypermobility Spectrum Disorders Support Community offered in partnership with The Ehlers-Danlos Society.* Inspire and BCH partnered with The Ehlers-Danlos Society to explore barriers to genomic medicine discussed in online community posts.

Methods

The dataset included 1,949 de-identified and anonymous posts from 232 members mentioning genomic medicine from the EDS and HSD Community, written between October 1, 2019 and September 30, 2021. While this project focused on EDS, individuals in our study group may instead have HSD due to the similarity of the two conditions.

Researchers used Inspire's Real World Voice (RWV) research methodology to identify and categorize posts about genomic medicine. Analysis of virtual, organic conversations from those with lived experience offer valuable insights into barriers to genomic testing, treatment, and counseling. A multidisciplinary team of researchers coded all posts mentioning barriers to genomic medicine and then for author type (e.g., patient, caregiver etc.), and barrier type using the modified social ecological framework.

Figure 1: Barriers to Genomic Medicine



Results

A total of 345 posts from the dataset mentioned barriers in accessing genomic medicine. The majority of barriers occur at the social-structural level (174) in the framework, followed by interpersonal (115); individual (81); and technological (14) (see Table 1).

Note: Totals for coded barriers exceed 345 because each could be coded for multiple barriers; 12 posts were removed from the dataset because they didn't provide enough information.

Table 1: Posts about genomic medicine in the EDS & HSD Community

	Barrier Type			
Author type	Social-structural n(%)	Interpersonal n(%)	Individual n(%)	Technological n(%)
Patient	154	99	65	11
Caregiver	17	16	9	2
Healthcare Provider	1	0	0	1
Unknown-Other	2	0	7	0
Total	174(50%)	115(33%)	81(23%)	14(4%)

Social-Structural barriers

Social-structural barriers include factors like societal norms, policies, and practices that influence health-related decision making and are outside the patient or caregiver's control, such as long wait times to see geneticists, excessive travel times, and high out-of-pocket costs (Kumar et al., 2012; SSDH, 2017). Solutions include availability and access to more virtual appointments, referrals/testing, and counseling.

Strategies from The Ehlers-Danlos Society

- Project ECHO program expands network of trained clinicians
- EDS and HSD conference scholarships address financial barriers
- Translation of EDS and HSD information and resources address language barriers

Experiences from the community

*"All of the genetic clinics within two hours from me refuse to diagnose EDS. The one that does [is] **2.5 hours away** has a wait list of 9 months."*

*"Unfortunately, because it's so **expensive and insurance does not want to pay for it**, many zebras, like myself, go without genetic testing at all."*

"I had never heard of EDS and waited a year to see a geneticist once I came across it."

Interpersonal barriers

Interpersonal relationships refer to the influence of family, friends, healthcare providers and others have on health and behaviors, such as family members who are unwilling to get genetic tests or be supportive for testing of loved ones; providers who are indifferent to patients or unwilling to give referrals, or help patients get genetic testing (Kumar et al., 2012). Solutions include improving communication among clinicians, patients, and family members.

Strategies from The Ehlers-Danlos Society

- Virtual support groups and Inspire community offer tips and best practices for resolving conflicts, strengthening relationships, and managing symptoms
- Project ECHO offers an "All teach, All Learn" model for clinicians to train other clinicians
- EDS and HSD Events and Scientific Meetings teach more clinicians about diagnosis and treatment

Experiences from the community

*"I had a geneticist diagnose me, but **she offered no further testing and not many treatment options**. I know there's not a lot of treatment...but she left me hanging."*

*"**A lot of geneticists are no longer diagnosing hEDS**--they say it's something you don't need a geneticist to diagnose, which may be true, but **very few other doctors feel competent to diagnose hEDS.**"*

Individual barriers

Individual barriers relate to knowledge, attitudes, behaviors, and beliefs that shape an individual's autonomy when engaging in health-related decision making, such as guilt about potentially passing on a genetic condition or confusion about the benefits of genomic medicine (Kumar et al., 2012). Solutions include dissemination of more health literate evidenced-based information about genomic medicine among patients, family members, and clinicians.

Strategies from The Ehlers-Danlos Society

- Research funding and dissemination of findings improves access to current scientific information (e.g., HEDGE Study)
- Project ECHO for community members trains individuals to advocate for their needs and have more effective medical appointments
- EDS and HSD Coalitions, facilitate collaboration advocacy (e.g., International EDS Consortium, Community Coalition, AHEAD Coalition, and Hypermobility Collaborative Network)

Experiences from the community

*"I didn't get it [the genetic test].... also, **what is it going to tell me? That I'm not crazy?"***

*"**I think geneticists only diagnose EDS. They do not typically treat it or offer support for symptoms.**"*

Technological barriers

Technological barriers include issues around software or hardware used by genomic medicine such as concerns about test accuracy, inconclusive results, and uninterpretable results (Newman et al., 2013). Solutions include:

- Availability of affordable genetic testing with easy-to-access results
- Access to professional counseling for increasing patient understanding and improving health outcomes
- Alternatives to in-person care with solutions for underserved populations (e.g. more virtual appointments and online registration; alternative options for individuals who face barriers with technology)

Strategies from The Ehlers-Danlos Society

- Individuals with technological and health literacy challenges can contact the EDS & HSD Helpline by phone or email for assistance for completing online forms, attending virtual appointments, and accessing patient portals

Experiences from the community

*"Does anyone know how to interpret raw genetic data to assess for different types of EDS? I searched online for specific SNPs to look up, but all I could find is the genes where different types of EDS stem from, but **nothing about how to specifically assess those genes.** Unfortunately, while there is very clear instructions on how to use your raw data to assess yourself for many diseases, **I can' find any similar instructions for EDS.**"*

Conclusion

Overall, patients reported experiencing social-structural barriers most frequently (>50%), followed by interpersonal, individual, and technological barriers. Efforts to integrate genomic medicine into clinical practices must address factors at all levels of the social ecological framework. Patients disproportionally face barriers that are outside of their immediate control, including interpersonal, technological, and social-structural barriers. Creating better access to genomic medicine requires policy and institutional changes to ensure that patients receive better and more comprehensive genetic diagnosis and treatment.

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*Ehlers-Danlos and Hypermobility Support Community shortened to EDS and HSD Community