Background, Needs and Expectations for Genetic Counseling in Adults with Ehlers-Danlos Syndrome

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BACKGROUND

Ehlers-Danlos Syndrome (EDS) is a heterogeneous group of heritable connective tissue disorders that are primarily characterized by joint hypermobility, tissue fragility, and skin hyperextensibility, in addition to a variety of other associated clinical features. The 2017 International Classification of the Ehlers-Danlos Syndromes now recognizes 13 subtypes. Hypermobile EDS (hEDS) is the most common subtype, but remains the only subtype with an unknown genetic etiology resulting in challenges for diagnosis. Unless the healthcare provider in question is knowledgeable of the wide range of symptomatology of connective tissue disorders, accurate diagnosis recognition is often delayed.

Although generalized joint hypermobility is the dominant clinical manifestation, the symptomatology spectrum of individuals with hEDS is complex with several studies reporting multisystemic involvement including fibromyalgia, chronic fatigue, joint/limb pain and gastrointestinal problems. In addition to the physical manifestations in hEDS, many patients report comorbid psychological manifestations. There is a high incidence of mental health disorders including anxiety disorders, depression, neurodevelopmental disorders, eating disorders and personality disorders (obsessive-compulsive) in this population.

Genetics professionals act as tertiary support for the ongoing care, coordination and management of patients with EDS. Genetic counselors and geneticists work regularly with individuals with a diagnosis of hEDS. However, poor understanding of hEDS leads to confusion in physician referrals and inappropriate care pathways. Without a known molecular etiology, and with complex manifestations, both physical and psychological, it is challenging for genetics professionals to know what to address in a genetics or genetic counseling session.

This exploratory study investigates the Background, Needs and Expectations (BNE) of genetic counseling in adults primarily with hEDS.

OBJECTIVES

1. Quantitatively explore the BNE of genetic counseling in individuals with EDS. Awareness of the BNE will aid healthcare providers, mainly genetic counselors, to better address the needs of this population by tailoring genetic counseling sessions, providing appropriate management and ultimately improving the quality of care.

2. Qualitatively assess EDS symptomatology, both physical and psychiatric, to assess the need for a multidisciplinary approach to care.

METHODS

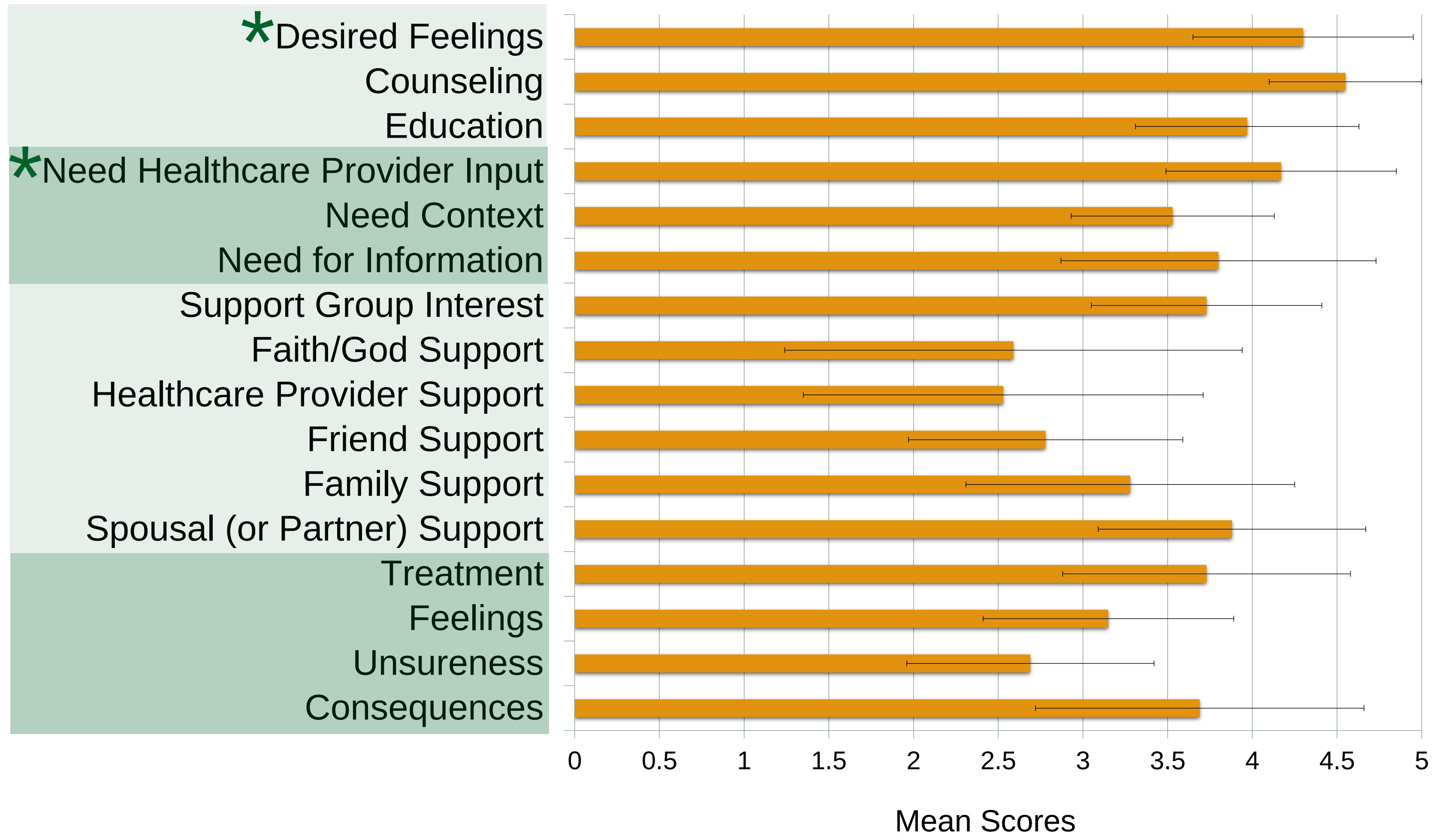
Adults (age 18 and over) with a clinical diagnosis of hEDS and/or molecular diagnosis of EDS were identified and recruited through the Columbia University Medical Center (CUMC) – Clinical Genetics patient registry from the past 10 years.

An online survey was created that included:

- A 61-item validated scale to assess and explore participants' self-reported BNE including beliefs about their genetic diagnosis, social support, need for information, and expectations for a genetic counseling session
- Questions investigating EDS diagnosis, any prior experience with a genetic counselor and experience with other doctors
- Qualitative open-ended questions that explored experiences with EDS by addressing the most disruptive symptoms, exploring psychiatric diagnoses, and inquiring into specialists involved in care and the need for multidisciplinary care

RESULTS

Fig. 1 Mean scores of the BNE scale subscales

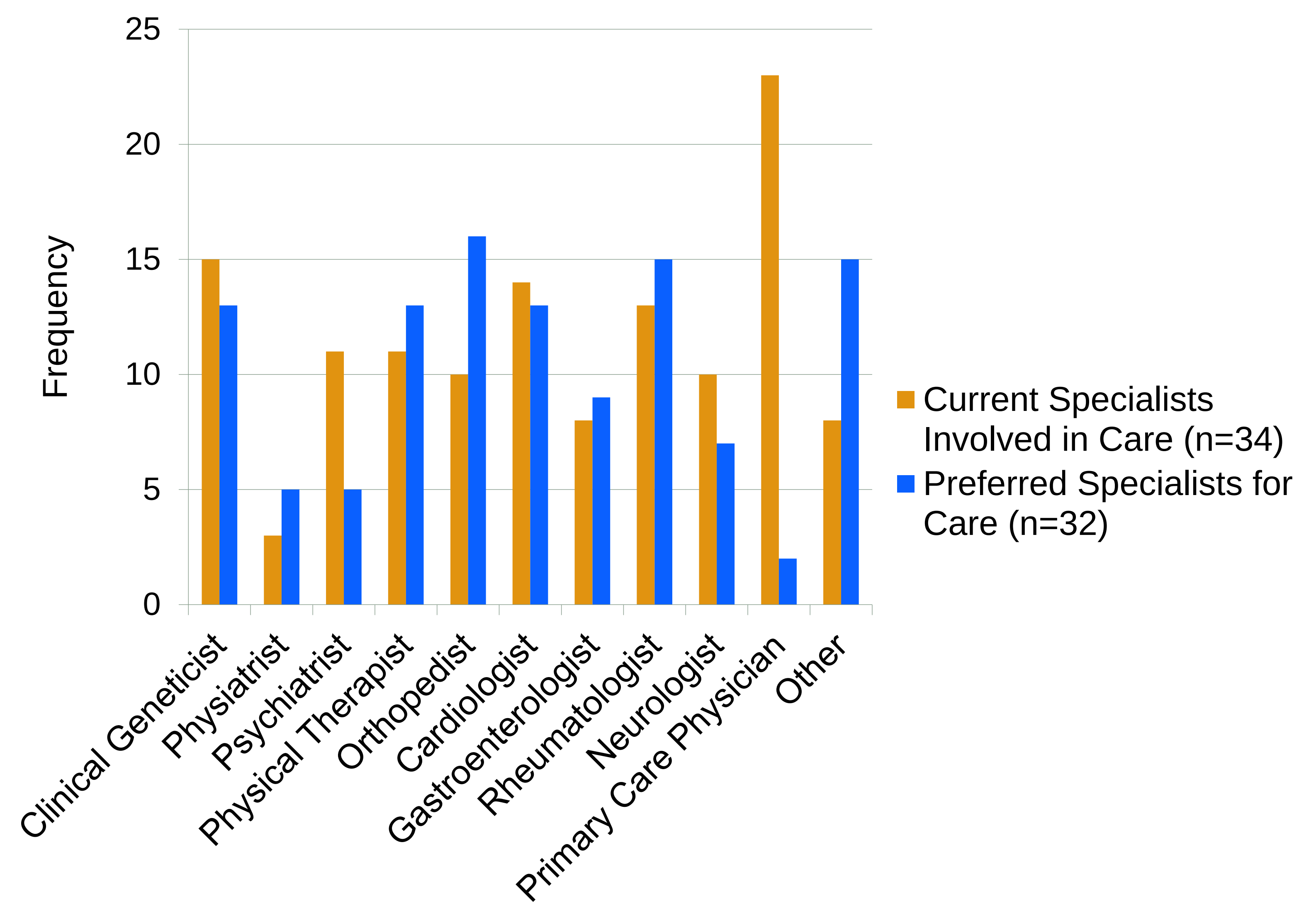


Scores range from 1 to 5, higher scores indicated stronger endorsement.

The 16 subscales presented above are grouped into 4 domains (Expectations, Needs, Social Support and Beliefs) that have been shown to influence the process and outcomes of a genetic counseling session. The internal reliability of the BNE subscales were greater than α > 0.7 with the exception of the Counseling subscale (α = 0.68). Descriptive statistics for each subscale based on the aggregate cohort have been reported in Figure 1 with standard deviation.

A one-way repeated measures within-subject ANOVA was conducted to compare the differences in mean scores of the Counseling subscale to all other subscales. Results indicated that the most significant subscales for this sample were the **Desired Feelings** subscale, F(1, 20)=55.67, p<0.01, and the **Need Healthcare Provider Input** subscale, F(1, 20)=44.25, p<0.01.

Fig. 2 Frequency of current specialists involved in care compared to respondents' preferred specialists for care



RESULTS

Table I. Clinical history of respondents

	N*	%
Age of Diagnosis (years)		
	29.0 + 11.4	
Clinical diagnosis of EDS		
Yes	30	81.00%
No	4	10.80%
Not sure	3	8.10%
Type of EDS		
Hypermobility type	23	82.10%
Classical type	4	14.30%
Not sure	1	3.60%
* The total study sample consists of 37 respondents.		

The physical symptoms of EDS reported to be the most disruptive to the individual's daily lives include **chronic pain** (76.4%), **gastrointestinal issues** (38.2%), **chronic fatigue** (29.4%) and **stress due to hyperflexbility** (23.5%).

The sample was split in incidence of psychiatric illness, with 45.7% reported having had experience with a psychiatric illness. Of these respondents, **depression and anxiety were common** (56.3% and 50.0%, respectively). Other psychiatric illnesses reported were **bipolar disorder** (12.5%) and Obsessive Compulsive Disorder, Post-Traumatic Stress Disorder, Attention Deficit Disorder, and Autism Spectrum Disorder (Other – 37.5%).

Table II. Respondents' experience with genetic counselors and doctors

	N*	%
Prior experience with a genetic counselor/genetics professional		
Yes	29	78.40%
No	7	18.90%
Not sure	1	2.70%
Experience with a genetic counselor/genetic counseling		
Extremely dissatisfied	0	0%
Dissatisfied	2	6.90%
Neutral - neither satisfied nor dissatisfied	4	13.80%
Satisfied	13	44.80%
Extremely satisfied	10	34.50%
Experience with doctors pertaining to EDS symptomatology		
Extremely well informed	1	2.90%
Somewhat well informed	14	40%
Not well informed	20	57.10%
* The total study sample consists of 37 respondents.		

CONCLUSIONS

This research study demonstrates that genetic counselors could better meet the needs and expectations of individuals diagnosed with hEDS by facilitating decision making related to their health care and health management. In addition, feeling *positive* following a genetic counseling session is highly desired. The findings of complex physical and psychiatric symptoms are consistent with results of previous literature, and warrant the need for multidisciplinary care. Healthcare providers, including genetic counselors, should be aware of the symptomatology and BNE for the EDS population in order to deliver enhanced, more consistent services.

REFERENCES

Castori, M., Morlino, S., Celletti, C., Celli, M., Morrone, A., Colombi, M., . . . Grammatico, P. (2012). Management of pain and fatigue in the joint hypermobility syndrome (a.k.a. Ehlers-Danlos syndrome, hypermobility type): Principles and proposal for a multidisciplinary approach. *Am. J. Med. Genet. Part A, 158A*(8), 2055-2070. doi:10.1002/ajmg.a.35483

Malfait F, Francomano C, Byers P, Belmont J, Berglund B, Black J, Bloom L, Bowen JM, Brady AF, Burrows NP, Castori M, Cohen H, Colombi M, Demirdas S, De Backer J, et al. (2017). The 2017 international classification of the Ehlers–Danlos syndromes. Am J Med Genet Part C Semin Med Genet 175C:8–26.

Peters, K. F., & Petrill, S. A. (2011). Development of a scale to assess the background, needs, and expectations of genetic counseling clients. *Am. J. Med. Genet Part A, 155*(4), 673-683. doi:10.1002/ajmg.a.33610