

**DAILY LIVING IN YOUNG ADULTS WITH EHLERS-
DANLOS SYNDROME (EDS)**

Honors Thesis

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Abstract

Background and Significance: Ehlers-Danlos Syndrome (EDS) is a rare, genetic connective tissue disorder that effects the production of collagen in the body, resulting in chronic pain, hypermobility and the need for many medical interventions. There is often a delay in diagnosis into adolescence or young adult life due to variability in multi-organ symptoms and their severity leading to misdiagnosis and uncertainty. Connective tissue holds the whole body together and when the collagen in that tissue doesn't work the way it is supposed to, it can make changes in the way people live their lives.

Methods: A qualitative, phenomenological study was conducted to explore what changes this condition can make to a person's life. 5 women aged 18-22 were interviewed in person or via video chat. These interviews were later transcribed and analyzed.

Results: Themes that emerged from the data were pain limiting daily tasks, the invisibility of the disease, and uncertainty about future of the condition.

Keywords: Ehlers Danlos Syndrome, connective tissue disorder, young adults, health related quality of life, lived experience, rare disease

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Introduction

Nurses care for all age populations and may see individuals with different diagnoses in a day. Most nurses will care for disorders that they have heard of or even treated before, but occasionally, they come across a patient with a diagnosis that they have never experienced. Rare diseases are often misdiagnosed and go undiagnosed for long periods of time. One rare disease that is no exception to delays in diagnosis is Ehlers-Danlos Syndrome or EDS. Ehlers-Danlos Syndrome is a connective tissue disorder that affects the entire body, as connective tissue is not only in joints, but it supports the skin, blood vessels, and all the tissues of the body (National Library of Medicine, 2017). This condition is genetic, and individuals may have no family history and go without a concrete diagnosis for years. Those with this condition struggle through chronic health issues that others would not but many people with EDS live completely normal lives while struggling with the complications.

Background and Significance

Worldwide, people are living with rare diseases. Rare diseases are harder to diagnose and often go misdiagnosed or undiagnosed. A study by Molster et al. (2016) surveying adults in Australia living with a rare disease found that 51.2% of respondents waited over a year for diagnosis with 30.0% waiting five or more years. Adults living with Ehlers-Danlos Syndrome participated at 6.4% more than any other diagnosis; 52 out of the total 810 respondents (Molster et al., 2016). People with EDS may undergo treatments inconsistent with their physical capability when the healthcare professionals caring for them lack knowledge of the condition (Berglund, Nordström, & Lützén, 2000). From a rehabilitation approach, there is limited information about the management of

EDS and often treatment fails (Celletti, Castori, Morico, & Camerotta, 2014). In one patient example, Celletti et al. (2014) suggested the use of repetitive muscle vibration to help with the feelings of chronic pain but more research needs to be done. Those living with EDS also have trouble with anesthesia not working as well as would be expected causing pain that unaffected individuals would not experience during procedures (Berglund et al., 2000).

Pain associated with Ehlers-Danlos Syndrome can also occur outside of procedures, many people with this disease live with chronic pain throughout their bodies (Berglund et al., 2000). This pain tends to be labeled by healthcare professionals without knowledge of the disease as psychosomatic leading to negative healthcare experiences (Berglund et al., 2000). Another study identified that those with EDS with noted possible anxiety and depression had complaints of back pain and tiredness (Berglund, Pettersson, Pigg, & Kristiansson, 2015). Anxiety levels were triple the depression levels in their study population (Berglund et al., 2015). Cederlöf et al. (2016) describe a greater risk of psychiatric disorders in those with Ehlers-Danlos Syndrome hypermobility type. That same study found that when those with EDS, hypermobility type were compared with siblings who do not have EDS, the risk for psychiatric disorders like ADHD, depression, and suicide attempts is doubled (Cederlöf et al., 2016). Earlier diagnosis of Ehlers-Danlos Syndrome, predicted fewer individuals who attempted suicide; this is thought to be from the comfort of having a reason for their condition (Cederlöf et al., 2016). People with EDS who work full time had a higher level of acceptance of their disability (Berglund, Mattiasson, & Nordstrom, 2003). Greater levels of acceptance of disability

are associated with a higher level of meaningfulness and better functional status (Berglund et al., 2003).

Methods

The aim of this study was to describe what daily life is like with Ehlers-Danlos Syndrome and was approved by the Institutional Review Board at Salem State University. After a thorough review of the literature, a questionnaire approach was chosen. A qualitative, phenomenological approach was chosen to capture the lived experience of the participants. A questionnaire was created with demographic and condition specific questions. Subjects were identified and asked to participate through snowball sampling and were given a disclosure statement, either electronically or physically, prior to being interviewed (LoBiondo-Wood & Haber, 2014). Participants were interviewed by the author in person or via video chat and their responses were recorded. Those recordings were later transcribed by the author to be analyzed. The transcriptions were then read through for similar themes. The perceived themes were written out and tallied so that those with three or more of the five participants discussing them became the main themes presented.

Results

Participant age ranges were 18-22 and all were female. Participants reported their own perceived severity of their condition as mild (n=2), moderate (n=3), or severe. One participant noted that their EDS was moderate but their comorbidities made it more severe and they were included into the moderate count.

Table 1. Demographics of participants (n=5)

| Participant | Gender | Age | Self-reported Severity of EDS |
|--------------------|---------------|------------|--------------------------------------|
| 1 | F | 18 | Mild |
| 2 | F | 22 | Moderate |
| 3 | F | 18 | Moderate |
| 4 | F | 21 | Moderate (Severe with comorbidities) |
| 5 | F | 18 | Mild |

Themes that came out of this research were:

- Pain limiting daily tasks,
- The invisibility of the disease, and
- Uncertainty about future of the condition.

Pain limiting daily tasks

The participants reported that pain limits their ability to do daily tasks that they would like to do. There was a consensus that most of them at one point or another have felt as if they could not make or keeps plans that they wanted to due to their pain. One participant noted, "...the pain with EDS is so unpredictable. You never really know what you're gonna wake up to or, you know, what you might feel like 5 minutes from now." Another participant spoke along the same lines and said, "I might have plans and I'm like I can't leave my bed or my house right now because I'll just fall apart if I take one step out the door." One participant talks about how even just wearing a backpack can cause subluxations and pain, "a lot of times the straps will, on certain backpacks, pull my shoulders out."

Another talks about how there is only so much that she can do in a day:

“Looking at normal people... they’ll go to school. They’ll go to the gym. They’ll go to work and they’ll hang out with friends after. It’s like with EDS you have to have your one thing for the day that’s important that day.”

Invisibility of the disease

Participants continually noted that their condition was unnoticeable to others.

Multiple participants discussed perception of their condition by others. One participant talked about knowing her own personal limits and how she feels when having to sit down when in public places like the mall:

“...if I’m going out with friends or something to like the mall...making sure I have a brace with me ...and having to sit down...thinking like being afraid people will think, ‘Oh she’s weird. Why does she have to sit down? She’s like this young able-bodied person.’”

Another talks about her experiences in an elevator:

“...sometimes I really have to take the elevator and... I’ve heard people say stuff about people getting off on the first floor, so I’m, like if I’m getting off on the second floor, well they’re certainly saying stuff about me because you can’t see... It is not a visible condition.”

Another participant talks about how hard it is to live with something that isn’t easy to see:

“It’s difficult to live with an invisible illness and I think for me it’s often a bit more visible than it is for some people... it’s a really weird feeling to be able to, you know, I can take off my neck brace or, you know, unhook the feeding pump and look like a totally normal human being, but I’m just as sick as I was 5 minutes ago...people definitely treat you differently...”

Uncertainty about the future of the condition

Participants overall seemed to be unsure of what their future holds.

One participant talked about watching her mom go through the complications cause by being treated improperly for EDS:

“the scary part is not wanting to end up like that... I was able to get the right treatment and not be put in bad situations where I was just gonna damage [my] body even more. So I think I’m worried but not to the point where I’m absolutely terrified.”

Another participant discusses her concerns about childbirth, “I’m quite concerned that I won’t be able to have children because my hips will separate.”

She also spoke about wondering about the efficacy of surgery for EDS:

“I’ve wondered about shoulder reconstruction surgery and hip reconstructive surgery, knee reconstructive surgery, but I don’t want to be a tin man... I wonder if that would actually help because of my ligaments are so stretchy that I wonder if they would jut stretch back out.”

Another talks about her surgical path and how there aren’t many others who have been through the same surgical path as she has:

“I think the future is very, sigh, uncharted... I’m probably like the third patient in the world who’s ever had this combination of fusion and um, venous stenting and that’s a bit nerve racking because we have absolutely no idea the longevity of this treatment. Um, we don’t know if it is going to fail 5 years down the line.”

She also discusses her own uncertainty:

“We don’t know if I will continue to need more surgery down the line and we just really have no idea. And, it’s so difficult for me to plan my future and decide what I want to do, knowing that I have no idea what my life will look like.”

Another participant discussed her concerns, “the biggest problem for me is I can deal with something as long as I know where it’s gonna go.” When asked about concerns for the future, one participant simply replied, “I’m not really thinking of the future.”

Discussion

Pain limiting daily tasks

Across all participants, there was a sense of limitation on everything from future jobs to just hanging out with friends. This limiting feeling comes from the pain associated with this disease and the complications. The chronic pain prevents participants from being able to do certain tasks that they want to do that those without this condition can.

Invisibility of the disease

Participants commented a lot on how hard it is to live with a disease that can leave them looking completely normal but give them so much pain and limitations in function. They discussed how they get treated differently than those who have more visible illnesses.

Uncertainty about future of the condition

Participants across the board commented on their future being up in the air and not really knowing exactly where they will progress. They all weren't really sure if the treatments they were getting would continue to work or if a different intervention would be needed all together down the line.

Discussion of Ancillary Data

One finding that could not be generalized to the whole population of people living with Ehlers-Danlos Syndrome but found in all participants' testimonies was strong family support. Specifically, every single participant mentioned their mothers as a major support person. This finding could be attributed to the age of the participants, as all were aged 18-22 and that is usually a time in a person's life that they may still live at home.

Although participants were uncertain about the future, a few of them were optimistic to see what the future and science would bring for discoveries. They talked about the hope for new treatments and possibility of a cure.

Part of each interview, participants were asked what they would tell someone who was newly diagnosed with EDS about EDS. Here are some words of advice from those with this condition.

“It is manageable and you can get on top of it and you should always put your body first and listen to your body... bracing is the most important thing I think and resting and just being okay with that and admitting to yourself that you might have weaknesses but you aren’t completely debilitated and you can still do things.”

“It’s normal I guess, well cause when you say that your hip’s out or your shoulder’s out people kind of look at you funny. They either look at you like what or you’re over reacting and I want to tell them that like no, I believe you. It’s real and it’s not normal for them, but it’s normal for us.”

“...take a deep breath... a diagnosis is just a word for what you’ve already been dealing with and it gives you a lot of power.”

“...being active is one of the most important things I think when it comes to a connective tissue disorder because your muscles are your best friends... finding a good relationship with a physical therapist is very important.”

“There’s more of us than you’d expect around here, just waiting for a new EDS friend. So you may think you are in the dark and you don’t, and nobody understands but it’s definitely more common than you’d think, just go on YouTube and people can also live fairly normal lives... just because you see one person... that doesn’t mean that’s your future.”

Nursing Implications

Nurses everywhere need to make themselves more aware of rare diseases that leave people appearing to be healthy, but who are in a severe amount of chronic pain. Nurses, especially those in emergency rooms and urgent care centers, who care for patients with frequent joint injuries should think about screening them for this condition. There is a need for more research about this condition, as there is not much out there about Ehlers-Danlos Syndrome. Nurses need to be educating those diagnosed with this condition about EDS and about the many ways to get emotional support.

Limitations

This was an unfunded study done by an undergraduate student as an honors thesis. With a little less than a year's time to complete this study, time was a major limitation. Taking that into consideration, the author was only able to use a convenience sample through snowball networking and had there been more time, there would have been a larger sample size. Though this study was open to men and women aged 18-30 with Ehlers-Danlos Syndrome, only women aged 18-22 were a part of the study. All of those in the study have the same rare disease and therefore these results cannot be generalized for others with different rare diseases. A few of the participants were known personally by the author which may prevent some participants from sharing as much as they would to someone who does not know them or the condition at all.

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Appendices

Daily Living in Young Adults with Ehlers-Danlos Syndrome (EDS) (Hollohan, 2017)

Questionnaire

1. How old are you?
2. What is your gender?
3. How old were you when you were diagnosed with Ehlers-Danlos Syndrome?
4. What type of Ehlers-Danlos Syndrome do you have?
5. Do you consider your EDS mild, moderate, or severe?
6. What systems are involved in your case of EDS? (cardiovascular, musculoskeletal, integumentary, gastrointestinal, ect.) Do you have any pain associated with EDS?
7. Do you have any other health conditions?
8. Do you know others with EDS? How do you know them?
9. How do you cope with EDS on a general day? How about on a bad day?

10. Who helps you with living with EDS?

11. Do you have trouble with activities that you need to do daily? Explain.

12. Are there things that you are not able to do because of EDS? What are they?

13. If you have no pain, skip this question. Would you say that the pain associated with EDS makes a difference in your day to day?

14. Does fatigue associated with EDS make a difference in your day to day activities?

15. Do you have any concerns about your future with EDS? Please explain.

16. What would you want to tell someone who is newly diagnosed with EDS about EDS?

17. Is there anything else that you would like or need to know about EDS? If so, do you have a resource to get that information?

18. Is there anything else you think I should know about living with EDS?