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The daily life and psychosocial impacts of Friedreich ataxia: A qualitative study of patient lived experiences

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1. Background & Objectives:

- **Friedreich ataxia (FA)** is a rare, progressive, and systemic neuromuscular disorder characterized by impaired motor function and speech, typically resulting in loss of ambulation.[1]
  - **Cardiomyopathy, diabetes mellitus, scoliosis** and **pes cavus** are common manifestations of the disease, and many patients experience **loss of vision** and **hearing**.[1]
  - The clinical course and rate of progression are variable and correlate with age at disease onset; **earlier onset** (<16 years of age) is indicative of **increased disease severity** and **reduced life expectancy**, with cardiac dysfunction being the most common cause of early mortality.[2]
- Impacts of FA on **patient health-related quality of life (HRQoL)** have been documented;[3] however, generic HRQoL measures can miss **key disease specific impacts**, particularly in rare conditions.
- **Qualitative methods** can provide unique insight into patient lived experiences by contextualizing the impact of clinical manifestations.
  - While several qualitative studies have documented challenges that FA patients experience related to diagnosis and disease management,[4,5] there is a **lack of research** to explore the **broader lived experiences** of patients with FA.

OBJECTIVE

To understand **patient perspectives** on the impact of FA symptoms on **daily life** and **psychosocial wellbeing.**

2. Methods

Participants	• Individuals with FA, or family caregivers reporting on their behalf, were recruited through patient advocacy groups in the United States (US) and Europe. • Eligible participants were 1) adults with FA or the primary caregivers of an adult or minor with FA; 2) living in the US, United Kingdom (UK), or Germany; 3) fluent speakers of English or German; and 4) capable of providing informed consent.
Data Collection	• Qualitative interviews were conducted using videoconferencing (Zoom) with audio recording; informed consent was obtained verbally. • Participants responded to open-ended questions about the impact of FA symptoms on daily life, including key challenges and adaptations. • Participants completed demographic and clinical questionnaires; results were used to classify interview data.
Analysis	• Interview content was transcribed; German interviews were translated into English. • Demographic and clinical characteristics were summarized. • Conventional content analysis[1,2] was used to explore patterns in lived experiences of FA clinical manifestations and symptoms, and their impact on patient HRQoL.
Ethical Review	• This study received approval from WCG IRB (Study number: 1336811)

3. Results:

- Mean (standard deviation) patient age was **33 (12.1)** years; **62.1%** were female and **55.2%** had symptom onset at <16 years of age (**Table 1**).
  - Of 29 participants, 13 were caregivers reporting on behalf of patients.
- **14 (48.3%)** patients were non-ambulatory; most non-ambulatory patients **(n=12)** reported symptom onset prior to age 16.
- All participants **(n=29)** reported problems with lower limb coordination and balance, and most **(n=28)** reported fine motor impairments.
  - Participants explained these symptoms lead to doing everything **slowly** and with **greater effort**.
- **22** participants reported **difficulties managing fatigue**, which was often described as one of the most challenging aspects of FA (**Figure 1**).
  - Fatigue limits participation; individuals often needed to carefully plan their activities to manage fatigue and would sometimes need to cancel plans with friends or family.
- **Emotional impacts** were common **(n=27)** and while many participants maintained a positive outlook much of the time, day-to-day situations could provoke feelings of frustration, anger, sadness, embarrassment or worry.
  - **Frustration** with physical limitations was frequently described.
  - **Loss of independence** was an emotional hurdle for many patients; this was closely linked to frustration with declining physical function.
  - Some described **feelings of worry** or **embarrassment** about how they are perceived by others.
- Despite challenges, individuals adapt to life with FA by finding ways to **maintain participation** and **social connection**, developing **coping strategies**, and identifying **sources of optimism** in their daily lives.
  - Strategies for self-care and coping included keeping busy to avoid dwelling on FA, practicing mindfulness or meditation, and taking control of one's health through diet and/or exercise.

Table 1. Patient demographics and clinical characteristics (n=29)

	All (n=29)	Patients (n=16)	Caregivers* (n=13)
Demographics			
Mean (SD) patient age at interview, years	33.0 (12.1)	37.8 (8.0)	27.2 (13.8)
Female gender, patient (n, %)	18 (62.1)	13 (81.3)	5 (38.5)
Country of residence (n, %)			
United Kingdom	10 (34.5)	6 (37.5)	4 (30.8)
Germany	9 (31.0)	5 (31.3)	4 (30.8)
United States	10 (34.5)	5 (31.3)	5 (38.5)
Patient clinical characteristics			
Age (years) at symptom onset (n, %)			
<16	16 (55.2)	7 (43.8)	9 (69.2)
16-25	9 (31.0)	6 (37.5)	3 (23.1)
>25	4 (13.8)	3 (18.8)	1 (7.7)
Time (years) since symptom onset (n, %)			
<10	5 (17.2)	1 (6.3)	4 (30.8)
10-19	12 (41.4)	7 (43.8)	5 (38.5)
20-29	10 (34.5)	7 (43.8)	3 (23.1)
30+	2 (6.9)	1 (6.3)	1 (7.7)
Loss of ambulation (n, %)	14 (48.3)	5 (31.3)	9 (69.2)

Abbreviation: SD, standard deviation

*Caregivers responded on behalf of the patient

Figure 1. Quotations from FA patient and their family caregivers illustrating key findings

Symptoms permeate everyday life: doing everything slowly

"His workday ends normally at 5, but he will continue throughout the night going to the computer and working on things, because it does take him longer to type." USCG4

"It takes me extra time to do anything… and of course I have to use expensive equipment to get around, but most of the time if I want to do it, I figure out a way to do it." USP5

Fatigue limits participation

"Fatigue's a huge problem. I mean, he's missed so much school because of it." UKCG3

"Fatigue is probably the worst thing that I deal with… Using a wheelchair is easy in comparison to managing fatigue. I'm quite good at it now, but yeah, it takes quite a long time to work out how to pace yourself." UKP6

Frustration with physical limitations and loss of independence

"He's very strong in terms of the long-term outlook, he doesn't sit and wallow… but the day-to-day frustration. So if he spills something, drops something, stumbles, falls… that really takes it out of him emotionally… "UKCG2

"He'll fall out of his chair… It's become an issue because he doesn't like to accept help. So I think that's one of the biggest problems, is being dependent on other people and coming to terms with that for himself is really hard." USCG4

Worry about how you are perceived

"It can be an embarrassing thing, really, having to use your hands and paying for things... Like, where do you carry things if you're in a wheelchair? So, it's a bit like an anxiety thing… it's just about not wanting people to sort of be looking at you and making a judgement." UKP3

"He hates being out and feeling like people are looking at him, which people just do because you sort of automatically do look at somebody in a wheelchair... I suppose all teenagers want to sort of disappear, or you don't want to stick out." UKCG4

Maintaining participation and connection

"She loves going to our boys' soccer and basketball games…We still go camping at least once every summer. We just find campgrounds that are as accessible as can be… We try not to let the Friedreich's Ataxia hold her back from doing things that she loves to do." USCG5

"I've met a lot of people that I would never have met had we both not had FA or had some connection to the FA community. Those people are very important to me." USP1

Self-care and coping strategies

"I mentioned about going to the gym and personal training, so I think that was almost my way of taking back control. So you can obviously work out and make yourself stronger and make yourself fitter, which clearly leads to better posture, better balance." UKP4

"Just staying busy, whether it's pedaling or staying busy with my daughter. I feel like when I'm busy that I'm happier because I don't have a whole lot of time to sit around and pout." USP4

Sources of optimism

"I've met a lot of people with FA and with a lot of the disabilities…I think it just makes you see the world in such a different way… everyone deals with something at some point, grief or whatever, and we all need to support one another. And I think FA has really helped me understand that." UKP6

4. Discussion & Conclusions:

- This study augments scarce data to describe the lived experiences of individuals with FA.
- Findings highlight the psychosocial challenges that permeate everyday life with FA, as well as sources of strength and adaptation.
- FA impacts participation in social activities, and day-to-day emotions fluctuate in response to physical limitations and the subsequent need to cope with loss of independence.
- FA patients demonstrate capacity to adapt to these challenges with support from friends and family members, identifying sources of optimism and strategies for coping and promoting social interaction and independence.
- Future quantitative studies to complement this research should include large, diverse samples to explore how HRQoL impacts correspond to key phases of the disease journey.

5. Limitations:

- Strengths of this study include: 1) the inclusion of a clinically diverse group of FA patients, with variation in both age at and time since disease onset, and 2) the collection of rich qualitative data to allow for a nuanced understanding of patient experiences.
- Study limitations are 1) that the perspectives of children with FA were not directly included, and 2) the potential for sampling bias.
- Parental caregiver reporting on behalf of children provided valuable insight into the experiences of young FA patients; however, research from other progressive conditions with childhood onset suggests that caregivers and patients may differ in the perceptions of HRQoL impacts.[1,2]
- Participants recruited through patient advocacy groups may not be representative of patients and caregivers who are not connected with, or who have less involvement, in these groups.

6. References:

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