PCR139

‘We have conversations that other couples don’t have to have’: A qualitative study of the impact of caregiving in Friedreich ataxia

SM Szabo, MSc;[1] A Bever, MPH;[1] D Lynch, MD PhD;[2] G Vasco, MD PhD;[3] P Giunti, MD PhD;[4] J Vallortigara, PhD;[4] I Tomazos, PhD MBA[5]

[1]Broadstreet HEOR, BC, Canada; [2]Division of Neurology, CHOP, PA, USA; [3]Department of Neurorehabilitation and Robotics, Bambino Gesù Children's Hospital, IRCCS, Italy; [4]Ataxia Centre, UCL Queen Square Institute of Neurology, London, UK; [5]PTC Therapeutics, NJ, USA

1. Background & Objectives:

- **Friedreich ataxia (FA)** is a rare, progressive and systemic neuromuscular disease characterized by impaired motor function and speech, typically resulting in loss of ambulation.[1]
- The clinical course and rate of progression are variable and correlate with age at disease onset, with **earlier onset** (<16 years of age) indicative of **increased disease severity** and potentially early cardiac-related mortality.[2] Other common manifestations may include scoliosis, diabetes mellitus, and impacted speech, vision and hearing.
- Because of the extent of **patient burden** and the **progressive manifestations** of FA, **caregiving** is typically required.
- There are limited data to describe the **health-related quality of life (HRQoL)** impacts of caring for somebody with FA;[3] and a recent review of caregivers in rare inherited diseases identified gaps in understanding **1)** mediators of burden, and **2)** how impacts differ between parental and spousal caregivers.[4]

OBJECTIVES

- This study aimed to understand the **impact of caring for those with FA on the daily life and HRQoL of spousal** and **parental caregivers.**
  - Secondary objectives explored **how experiences compared** between **spousal** and **parental** caregivers.

2. Methods

Participants
- Family caregivers of individuals with FA were recruited through patient advocacy groups in the United States (US) and Europe.
- Eligible participants were (1) 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 were asked to describe the impact of caregiving on daily activities and emotional outlook, including challenges and strategies for mitigating these; prompts were open ended.
- Caregivers completed demographic and clinical questionnaires; results were summarized and used to classify interview data.

Analysis
- Interview content was transcribed; German interviews were translated into English.
- Demographic and patient clinical characteristics were summarized.
- Conventional content analysis[1,2] was used to explore patterns in participant responses.

Ethical Review
- This study received approval from WCG IRB (Study number: 1336811)

3. Results:

- Among 13 caregivers, the mean (standard deviation) age was **43.2 (5.9)** years; **76.9%** were female and **69.2%** cared for patients with symptom onset at <16 years of age (**Table 1**).
- Caregivers reported varied impacts on daily life and HRQoL due to caregiving (**Figure 1**).
- All **13** participants described ‘making space’ for caregiving, which impacts **ability to work**, **socialize**, and maintain a **healthy lifestyle**.
  - FA affects the whole family; caregivers spoke about how, within the family unit, everyone’s life is dictated by routine.
  - Relationships with friends and family change with FA.
- **Emotional stressors** were substantial and demands ever-present; **frustration** and **worry** were common.
  - Some caregivers describe caregiving as **grueling**, and worry about suffering **burnout**.
  - Caregivers describe **worry** and **sadness** about the inevitable progression their loved one will face, worry that they personally should have done more, and the stress of a disease without a cure.
- **6** caregivers highlighted sources of positivity like increased **empathy** and strengthening of **relationships**.
  - Many reiterated how critical it was to them that FA does not define them, or their families.
- **9** described **coping strategies** including being available for themselves, mindfulness, and participation in research.
- Perceived **caregiving burden** varied based on effectiveness of coping strategies, intensity of demands, availability of external support, and other personal factors.
- **Spousal caregivers (n=7)** discussed unique challenges around family planning and intimacy, needing to shoulder more household tasks, or having to consider the impact of premature mortality.
  - At the same time, spousal caregivers often noted that they and their partner have a level of closeness they did not think they would have had, without FA.
- Some **parental caregivers** discussed how the stress of the diagnosis and caregiving demands threatened their adult relationships; for other parents, they reported caregiving brought them closer together.

Table 1. Caregiver and patient characteristics (n=13)

Demographic characteristics		
Mean (SD) caregiver age at interview, years		43.2 (5.9)
Mean (SD) patient age at interview, years		27.2 (13.8)
Female gender, caregiver (n, %)		10 (76.9)
Female gender, patient (n, %)		5 (38.5)
Caregiver type (n, %)	Spousal caregiver	7 (53.8)
	Parental caregiver	6 (46.2)
Country of residence (n, %)	United Kingdom	4 (30.8)
	Germany	4 (30.8)
	United States	5 (38.5)
Patient clinical characteristics		
Age (years) at symptom onset (n, %)	<16	9 (69.2)
	16+	4 (30.8)
Time (years) since symptom onset (n, %)	<10	4 (30.8)
	10-19	5 (38.5)
	20+	4 (30.8)

Abbreviation: SD, standard deviation

Figure 1. Quotations from FA family caregivers illustrating key findings

Caregivers and families need to make space in their lives to care for their loved one with FA

“I’ve **changed my working environment** so that I’m a lot more flexible... it works better for [husband’s name] because I was **available for a lot more of his appointments**... I learnt to work that into my daily life.” UKCG5

“**Some friendships became even deeper** because of the diagnosis whereas **other friendships became looser**.” DECG7

Emotional stresses are substantial and ever-present

“Yeah, **it can be grueling**…**Love doing it**. But a lot’s changed in the last five years and it seems the change is maybe… noticeable every few months. I go, ‘Wow, we can’t do this anymore.’ But it is a little bit… **it’s exhausting**. USCG3

“Obviously **it’s challenging** to watch your child lose abilities over time, their condition worsen. One thing I always think to myself every night when the girls are going to bed and kiss them goodnight, that there may be a day that my wife and I are still here and they’re not because of this disease. **How do you cope with that**?” USCG2

Emotional stresses are balanced by positive aspects of caregiving

“It’s created a really tight unit. Like **my boys will do absolutely anything for their mother**… and if we don’t feel like doing the mundane thing, like playing boardgames, but we know that’s what mama wants to do, because it’s spending time together, we just bite the bullet and do it, because it’s going to make her happy.” USCG5

Individualized coping strategies

“The moment is now; today is the day we are living. W**e don’t look into the future so much** because it may be scary to think about the things that may be in store for us. And we don’t want to focus too much on the condition because **every person with FA has a certain share of healthiness** in them…. We should not define the person just by their condition.” DECG8

“There’s always this **silver lining** that some of our closest, closest in the world are others that have the same disease, so there’s a **sense of community**. Once a year we go to an event… a big symposium, and it’s like a family reunion.” USCG3

Differences observed between parental and spousal caregivers

“**We have conversations that other couples don’t have** to have about a lot of things…And we’ve had a lot of talks ourselves about how to prepare, how we want to prepare them, and how we think they would best feel supported, and how to give them experiences that they can hold on to so that they still feel connected to their dad.” USCG4, spousal caregiver

“I’ve split up from his dad, but **I think his dad struggles** with it. **He doesn’t have a lot of contact with [child’s name] anymore**. I just think he just can’t deal with it when we got the diagnosis... Yeah, I mean, that’s difficult.” UKCG3, parental caregiver

4. Discussion & Conclusions:

- This study contributes novel insights into the impact of FA on both caregivers and the broader family unit.
- While the impact of caregiving on emotional health, daily activities, and relationships can be substantial, these can be balanced by sometimes surprising positive aspects.
- Perceptions of caregiving intensity, which are multifactorial, can vary substantially, and differ between spousal and parental caregivers.
- Important determinants of caregiver impact identified include availability of external supports, personal characteristics and coping strategies of the caregiver, and the caregiver’s relationship to the patient.
- These findings augment scarce data on the impact of caregiving in FA.

5. Limitations:

- The inclusion of both parental and spousal caregivers was an important strength of this study; findings demonstrate that experiences and perceptions of caregiving for a child with FA differ substantially from caring for a spouse.
- A limitation of this study is the potential sampling bias resulting from recruitment through patient advocacy groups; caregivers who are highly involved in these groups may benefit from greater access to external support networks or other resources that impact perceptions of caregiving ‘burden’.
- Future research may wish to expand on these findings by using quantitative methods to assess caregiver impact in a larger sample of individuals with experience of caring for a family member with FA.

6. References:

1. Cook A & Giunti P. Br Med Bull. 2017 Dec 1;124(1):19-30. **2**. Parkinson MH et al. J Neurochem. 2013 Aug;126 Suppl 1:103-17. **3.** Buesch K & Zhang R. Curr Med Res Opin. 2022 Oct; 38(10):1739-1749**. 4.** Hsieh H-F & Shannon SE. Qual. Health Res. 2005;15(9):1277-1288. **5**. Vaismoradi M et al. Nurs Health Sci. 2013 Sep;15(3):398-405.

FUNDING & DISCLOSURES: This study was funded by PTC Therapeutics. SMS and AB are employees of Broadstreet HEOR, which received funding from PTC Therapeutics for this work. GV, PG, and JV received consulting fees from PTC Therapeutics. IT is an employee of PTC Therapeutics. Medical writing and editorial support was provided by Broadstreet HEOR and was funded by PTC Therapeutics.

CONTACT INFORMATION: Ioannis Tomazos, ytomazos@ptcbio.com

THE INTERNATIONAL SOCIETY FOR PHARMACOECONOMICS AND OUTCOMES RESEARCH (ISPOR)
EUROPEAN CONFERENCE, COPENHAGEN, DENMARK, NOVEMBER 12-15, 2023