Exploring Patient and Caregiver Perspectives on Burden Associated with Friedreich Ataxia in the USA

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Objective

Friedreich ataxia (FA) is a hereditary degenerative disease with clinical manifestations in multiple organs characterized by progressive gait and limb ataxia. Qualitative interviews were conducted with patients and caregivers to characterize the burden experienced related to FA symptoms and impacts.

Conclusions

Results show that from both the patient and caregiver perspective, numerous symptoms are experienced by people living with FA. FA is highly burdensome for patients & caregivers, negatively impacting their daily lives and limiting their ability to lead a functional life, in areas like mobility, self-care, and productivity, as well as imposing a financial burden.

● Patients (N=20)

● Caregivers (N=11)

Methods

Study Design

- A sample of 20 FA patients and 11 caregivers were recruited from the Friedreich's Ataxia Research Alliance (FARA) registry and FA Facebook communities.
- Individual qualitative interviews were conducted via telephone by trained qualitative research interviewers.
- Interviews lasted 45 minutes, were audio recorded and transcribed for qualitative analysis.
- Transcripts were coded using ATLAS.ti[1] software to group concepts by similarity of content (theme).
- The data on symptoms and impacts were grouped into sub domains by the authors to enhance understanding of the data.

Table 1. Characteristics of Interview Population

Characteristics	Patients (N=20)	Caregivers (N=11)
Current age in years Mean (SD) [Range]	28.5 (4.6) [22-36]	49.6 (8.4) [32-64]
Female (n (%))	15 (75%)	8 (73%)
Working full-time (n (%))	7 (35%)	9 (81.8%)
Total # of children Mean (SD) [Range]	Not asked	1.9 (0.6) [1-3]
# of children diagnosed with FA Mean (SD) [Range]	Not asked	1.3 (0.5) [1-2]
Sought genetic test based on symptoms (n (%))	Not reported by patients	11 (100%)
Age when diagnosed with FA Mean (SD) [Range]	15.15 (6.40) [5-27]	N/A

SD=standard deviation; N/A=not applicable

Results

- The 20 patients interviewed ranged from 22 to 36 years of age (mean 28.5). 75% were female and 7 (35%) were working full-time (Table 1).
- The caregivers provided an average of 55.5 hours of care per week .
- The most frequently reported symptoms by patients were neurological including impaired speech (n=14; 70%), loss of balance (n=10; 50%) and decreased muscle coordination (n=8; 40%) (Table 2).
- Caregivers most frequently observed the following symptoms to the patient; loss of balance, tiredness (each reported by n=6; 55%), and decreased muscle coordination (n=4; 36%) (Table 2).
- Patients rated their symptom bothersomeness at 6.7, impact of FA on their ability to live their daily lives at 7.1, and their financial burden due to FA at 6.5, all on a 9-point scale where higher scores indicate worse outcomes.
- Caregivers observed more symptom bothersomeness (7.4), lower impact on daily lives (5.8) and financial burden (5.3) to the patient (Table 3).
- Patient-reported negative impacts of symptoms included limited function in activities of daily living such as walking (n=15; 75%), dressing (n=11; 55%), limited social activities (n=11; 55%) and falls (n=6; 30%) (Table 4).
- Additionally, 91% of caregivers reported observing falls, and 73% reported issues with patient personal care and hygiene.

Table 2. Symptoms Associated with FA

Sub-domain	Symptom Description	Total Reporting Symptom; N (%)	Total Observing Symptom; N (%)
Neurological	Impaired Speech	14 (70%)	3 (27%)
	Loss of Balance	10 (50%)	6 (55%)
	Muscle Coordination Decrease	8 (40%)	4 (36%)
	Muscle Weakness	5 (25%)	2 (18%)
	Difficulty Swallowing	5 (25%)	1 (9%)
	Hearing Loss	4 (20%)	1 (9%)
	Vision Loss	4 (20%)	1 (9%)
	Unsteady Movements	3 (15%)	1 (9%)
	Loss of Proprioception	3 (15%)	1 (9%)
	Muscle Spasms/cramps	2 (10%)	1 (9%)
Energy-Related	Fatigue	9 (45%)	2 (18%)
	Tiredness	6 (30%)	6 (55%)
	Weakness	2 (10%)	0 (0%)
Genito-urinary	Urinary Urgency	3 (15%)	0 (0%)
Body – Other Symptoms	Development of Comorbidities (cardiomyopathy, diabetes)	6 (30%)	4 (36%)
	Scoliosis (spine curvature)	5 (25%)	4 (36%)

Table 3. Ratings Regarding Challenges with FA

Rating of Symptom Bothersomeness, Impact of FA on Daily Life, and Financial Burden Mean [Median]	Patients N=20	Caregivers N=11
How **bothersome are those symptoms** to you/them in your/their daily life? (scale of 1-9 with 1=not bothersome and 9=extremely bothersome)	6.7 [7]*	7.4 [8]^
On a scale of 1 to 9 where 1= no impact at all and 9= extremely high impact, how would you describe the **impact of your FA** on your/the person's **day-to-day life**?	7.1 [8]^	5.8 [5]¥
On a scale of 1 to 9 where 1 = no burden at all and 9 = extremely high burden, how would you describe your **current financial burden** associated with FA?	6.5 [7]Ϫ	5.3 [5]Ϫ

* n=2 not asked/or did not specify a rating; ^ n=3 not asked/or did not specify a rating; ¥ n=6 not asked/or did not specify a rating; Ϫ n=4 not asked/or did not specify a rating

Figure 1. Patient & Caregiver Quotations

It is really sad to see a decline in his mobility, like, week by week. (Caregiver)

The end of Friedreich's is not good. So I get pretty depressed whenever I think about it. (Patient)

I cannot pull up my pants on my own… he helps me stand up and pull my pants up. (Patient)

Some days he may fall more than others for whatever reason. (Caregiver)

Table 4. Impact of FA on Daily Life

Sub-domain	Impact Description	Total Reporting Symptom N (%)	Total Observing Symptom N (%)
Changes in Daily Performance and Activities	Dressing	11 (55%)	6 (55%)
	Personal Care/Hygiene	8 (40%)	8 (73%)
	Food Preparation	8 (40%)	3 (27%)
	Household Tasks/Chores	7 (35%)	1 (9%)
	Eating/Drinking (cutting food/handling utensils)	6 (30%)	7 (64%)
	Daily Activities	5 (25%)	2 (18%)
	Driving	4 (20%)	5 (45%)
	Reliance on Others	4 (20%)	0 (0%)
	Awareness/Caution in Activities	3 (15%)	3 (27%)
	Lives with Family	3 (15%)	2 (18%)
Impacts to Walking	Difficulty Walking	15 (75%)	10 (91%)
	Use of Wheelchair	13 (65%)	7 (64%)
	Use of Walker	11 (55%)	6 (55%)
Impacts to Mobility (Besides Walking)	Difficulty Running	8 (40%)	3 (27%)
	Difficulty with Stairs	4 (20%)	2 (18%)
	Difficulty Standing	2 (10%)	5 (45%)
	Difficulty Transferring	2 (10%)	2 (18%)
Falls	Falls	6 (30%)	10 (91%)
	Injuries Due to Falls	3 (15%)	3 (27%)
Relationship and Social Impacts	Social Life Limited	11 (55%)	7 (64%)
Emotional Functioning	Irritation/Annoyance	7 (35%)	0 (0%)
	Fear	4 (20%)	2 (18%)
	Anxiety	3 (15%)	4 (36%)
	Frustration	3 (15%)	4 (36%)
	Depression/Sadness	3 (15%)	3 (27%)
	Stubbornness	2 (10%)	2 (18%)
School/work impacts	Losing Employment/Employment Opportunities	6 (30%)	2 (18%)
	School Work/Studying (reduced ability to complete)	3 (15%)	9 (82%)
	Missing School/Work	3 (15%)	5 (45%)
	Reduced Ability to Work Outside Home	3 (15%)	3 (27%)
Additional Impacts	Choking	6 (30%)	1 (9%)
	Difficulty Writing	2 (10%)	2 (18%)

Limitations & Conclusions

- Patients and caregivers voluntarily contributed their time to this research. This evidence is generated from a convenience sample that may impact the generalizability of this research to the broader FA population. Despite this limitation, this evidence is very valuable, since it is the first published evidence elicited from the personal perspective of FA patients and caregivers.

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Disclosures

None

References: 1. Friese S. *User's Manual for ATLAS.ti 7.1.0*. 2013. https://atlasti.com/manuals-and-documents

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