Friedreich's ataxia patient pathway in Europe

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Background

Friedreich's ataxia (FA) is a rare progressive and multi systemic neurodegenerative disorder characterized by loss of coordination, typically resulting in loss of ambulation[1]. Cardiomyopathy, diabetes mellitus, and scoliosis are common and serious manifestations of the disease. Speech is impacted, and many patients experience loss of vision and hearing.

FA affects ~1:20,000 lives births in western Europe, where it is the most prevalent form of inherited ataxia[2]. Clinical course and rate of progression are variable and are influenced by age at disease onset[3]. FA typically presents between 5 and 15 years of age; these patients have a shortened life expectancy, with the most common cause of death being cardiac dysfunction.

While some 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 and care pathway of FA patients in different countries and navigating different health systems. In this study, we collected data from patients living with FA in three different countries, UK, Germany and Italy, to look at their care pathway, resource use and feedback about healthcare visits, treatments and care received, and gaps to be addressed to improve their care. In addition, we explored the role of specialist ataxia services (SAC) for the diagnosis and management of Friedreich's ataxia in selected countries.

Methods

We collected data between 2019-2021 in the UK, Germany and Italy using a patient survey of numerous questions, to gather information about the diagnosis and the management of the ataxias in specialist (SAC) and non-specialist settings, utilisation of other primary and secondary health care services, and patients' satisfaction of received treatment. At the time of the survey distribution, we counted two centres in the UK, nine in Germany and eleven in Italy, all spread out across respective countries. Patient organisations of each country, as well as clinicians in SACs in both Germany and Italy, were involved in the dissemination of the survey. The inclusion criteria were a confirmed diagnosis of ataxia, aged minimum 16 years old, and living in the selected country. We compared responses between SAC and non-SAC groups where appropriate and used Chi-square as statistical test.

References: 1. Cook A, Giunti P. , Br Med Bull. 2017 Dec 1;124(1):19-30.; **2.** Vankan P. , J Neurochem 2013;126:11–20.; **3.** Parkinson M.H. et al., J Neurochem. 2013 Aug;126 Suppl 1:103-17.; **4.** Daker-White G. et al, SAGE open medicine. 2013;1:205031211350556O. 5. Maring J. et al, Pediatric physical therapy: the official publication of the Section on Pediatrics of the American Physical Therapy Association. 2013;25(3):305.

Acknowledgements

This study was sponsored by the EBC as part of the Value of Treatment project, Takeda Pharmaceuticals, Reata Pharmaceuticals / Biogen and the Charity Friedreich Ataxie Förderverein e.V.

Results

Results - Sample characteristics

Participants: 27 from the UK, 14 from Germany and 56 from Italy

Characteristic	UK	Germany	Italy
Respondent type			
Patient	20 (74.1)	11 (78.6)	45 (80.4)
On behalf of the patient	7 (25.9)	3 (21.4)	11 (19.6)
Age (years)			
16-29	6 (24)	9 (64.3)	11 (19.6)
30-59	15 (60)	5 (35.7)	43 (76.8)
60-79	4 (16)	0 (0)	2 (3.6)
80+	0 (0)	0 (0)	0 (0)
Sex			
Female	17 (68)	5 (35.7)	36 (64.3)
Male	8 (32)	9 (64.3)	20 (35.7)
Attendance at SAC			
Never been	12 (66.7)	0 (0)	5 (11.1)
Currently going	3 (16.65)	12 (85.7)	30 (66.7)
Used to go	3 (19.4)	2 (14.3)	10 (22.2)

Figures are numbers in each category (%).

Results - Diagnosis

- **Most participants received their diagnosis from a neurologist in all three countries.**
- **in Germany (57.2%) and Italy (60%) patients saw a neurologist within 6 months of seeking medical advice - sooner compared to the UK (32%).**
- **Half of participants in Italy went straight to a SAC for that referral to see a neurologist versus 15% in Germany (no data for the UK).**
- **37% of people in Germany got their diagnosis in SAC / 60% in Italy so timely referral to a SAC could play a role in getting a diagnosis sooner.**

Referral to SAC

N (%)	UK	Germany	Italy
GP	2 (30)	4 (28.6)	8 (21.6)
Neurologist	3 (50)	6 (42.9)	16 (43.3)
Other	1 (20)	4 (28.6)	6 (16.2)
Unsure	0 (0)	0 (0)	7 (18.9)
Total	6 (100)	14 (100)	37 (100)

Other in UK: research study at a SAC; Other in Germany: research study, social-paediatrist center, self-referral; Other in Italy: self-referral, orthopaedist, psychiatrist, physiatrist.

Results – Multidisciplinary team (MDT) clinic

- **Attendance to MDT: UK 58%, Germany 33%, Italy 53%.**
- **Referral to an MDT in Germany: 75% by GP and 25% by non- SAC neurologist; in Italy: 38% by non-SAC neurologist and 33.3% by SAC neurologist, others by self-referrals, physiatrist, physiotherapist, Charity; no data for the UK.**
- **Effectiveness of the MDT care delivered: 80% positive in UK, 75% in Germany and 82% in Italy.**

Results – SAC access and retention

Reasons selected	UK	Germany	Italy
Never went to SAC	Problems with travelling (4/8) Equal care locally (3/8) Lack of awareness about SAC referral (1/8)	–	Did not wish to be referred (2/5) Referral not offered (2/5) Unsure why (1/5)
Stopped going to a SAC	Problems with travelling (2/3) Service not useful (1/3)	Reviewed cancelled (COVID) (1 response)	Problems with travelling (2/9) Service not useful (2) Equal care locally (3) Transfer to another service (1/9)

Results – SAC service

% positive feedback	UK	Germany	Italy
Coordinating referrals to HCPs	100	46.1	51.4
Offers to take part in research	100	84.6	78.4
Liaising with social workers	66.7	58.3	48.7

This is the feedback from participants who currently attend a SAC and those who used to attend such services.

Feedback on symptoms management and personalised care:

- ***UK: no difference of feedback from SAC versus non-SAC group***
- ***Germany: SAC group only with 89% positive feedback for both aspects***
- ***Italy: 100% positive feedback from non-SAC and 68% from SAC group for symptoms management; 33% in non-SAC and 59% in SAC group for personalised care***
- ***Overall, 77% in Germany and 51.5% in Italy found the care received in SAC was better than non-SAC services (data not available for the UK).***

Results – Resource use (top numbers of visits per speciality)

Most common visits	UK	Germany	Italy
SAC group	Other consultants (5) OT (4) Physiotherapist (4) Neurologist (3.3) SAC (1)	Physiotherapist (22.3) OT (15.6) GP (5.3) SLT (4.7)	Physiotherapist (19) SLT (5.6) Other consultants (1.7) OT (1.4)
Non-SAC	OT (2.7) Physiotherapy (2.5) Neurologist (2.4) GP (2.4)	–	Physiotherapy (23.3) OT (5) SLT (4) Other consultants (1.5)

Numbers are the mean number of visits per patient per year. Visit to neurologist: non-SAC. Other consultants: specialists other than neurologists; OT: Occupational therapist; SLT: speech and language therapy.

Health Economic Evaluation summary:

- ***Numbers of contacts for the different types of health service use between the SAC and non-SAC groups were non-significant.***
- ***non-SAC group show differences between countries: physiotherapy visits (the highest number of mean visits per patient per year were in Italy, followed by the UK, P=0.02).***
- ***SAC group show differences between countries: neurologist outpatient visits (the highest number of visits were in the UK, then Germany, then Italy, P=0.02).***
- ***No significant differences in mean total costs per patients per year between non-SAC and SAC group.***
- ***UK: costs are 4664€ per patient per year for non-SAC group versus 829€ for the SAC; Germany: 5604€ for the SAC; Italy: 958€ per patient per year for non-SAC group versus 963€ for the SAC.***
- ***Modal travel time to an SAC was 1-2 hours for UK and Italy and less than 1 hour for Germany, with a small proportion of patients travelled for more than 2 hours in all 3 countries.***
- ***For both Non-SAC, and SAC patients the modal travel time to see the neurologist at the general neurology clinic was less than 1 hour.***
- ***Most common mode of transport to the SAC and non-SAC clinic was via car***

Conclusions

The aim of this study was to get a better understanding of the Friedreich's ataxia patients' pathway in different European countries. We focus on a diagnosis, treatment and clinical care to manage the condition. The survey has highlighted:

- Patients' appreciation for SAC services compared to non-SAC, without any significant change in the costs.
- We identified difficulties in attendance and adherence, being physical distance to centres, and a lack of awareness about SACs.
- Data show that the visit to an MDT clinic was effective for FA patients.