

HEALTHCARE AND ECONOMIC IMPACT OF FRIEDREICH ATAXIA IN ITALY: A STRATIFIED ANALYSIS BASED ON PATIENTS' AMBULATORY STATUS

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Objective

This study aimed to assess Friedreich Ataxia (FA) patient resource utilization in Italy, based on their mobility status: ambulatory vs non-ambulatory, from the Italian National Healthcare Service (NHS) and Societal perspectives.

Conclusions

Loss of ambulation is associated with increased costs from both NHS (+€ 54.726) and societal (+€ 101.764) perspectives.

Developing new therapeutic options could slow the progression of the disease, support patients' independence, and reduce healthcare service and societal costs.

Introduction

- Friedreich Ataxia (FA) is a rare, inherited, and progressively neurodegenerative disorder that primarily affects patients' mobility, spasticity and cardiac function
 - FA is caused by mutations in the *FXN* gene, resulting in reduced levels of frataxin, a mitochondrial protein essential for cellular energy production.
 - Onset typically occurs during childhood or adolescence and leads to a gradual loss of voluntary motor coordination, impaired mobility, speech difficulties, and increasing dependence on others for basic daily activities. These symptoms contribute to a significantly reduced life expectancy and quality of life.
 - As the disease advances, patients may also develop cardiomyopathy, diabetes, vision and hearing impairments, and further communication challenges.
- Due to its multisystemic nature, FA imposes a considerable clinical and economic burden on patients, caregivers, and healthcare services, underscoring the urgent need for targeted therapeutic strategies and efficient resource allocation.

Methods

- Data from a multidisciplinary group of experts consisting of neurologist, 1 physiatrist, 1 cardiologist experienced in the treatment of FA were collected.
- Patients were classified as ambulatory or non-ambulatory based on their score in the walking/falling questions in the "FA-Activities-of-Daily-Living" (FA-ADL) scale: scores of 0-2 indicated ambulatory, scores of 3 or 4 indicated non-ambulatory status.
- Resource use was estimated across five FA-ADL composites (walking/falling, dressing/hygiene/feeding, swallowing/speech, bladder function, sitting position).
- Prevalence and resource use of patients with cardiomyopathy and scoliosis were estimated.
- The typical resource used by patients in each score across the 5 FA-ADL domains were estimated, via an ad hoc questionnaire, based on expert knowledge.
- Annual (e.g., specialist visits, formal/informal care, home health nursing, integrated home care (ADI), physiotherapy, emergency services) and one-time (i.e., Intensive Care Unit (ICU) hospitalization, pacemaker implantation, wheelchair, Hoyer lift, speech devices, home/car modifications) resource use were estimated and adjusted for disease severity in other domains and complication prevalence to determine incremental needs (based on expert opinion) (Table 1).
- Costs were calculated using Italian NHS tariffs for healthcare resources, literature or regional/national tariff for non-medical resources (Table 1).
- Costs were expressed in €/patient/year by multiplying the proportion of users, frequency, and unit cost for ambulatory and non-ambulatory groups.

Resource Use	Unit Costs	Annual Frequency	
		Ambulatory	Non-Ambulatory
N° of specialist visits	€16.20	8.1	25.8
ICU + Hospitalization for pacemaker	€19,052.00	0.0	one-time
Days of formal caregiver	€62.91	30.0	272.5
N° of home health nurse	€28.00	30.0	272.5
Days of informal care	€116.00	121.3	365.3
ADI (N° of months)	€1,269.00	0.0	12.0
Cardiomyopathy	€6,303.70	0.2	0.5
N° of emergency department visits	€350.00	0.5	1.0
Wheelchairs	€2,843.52	0.0	one-time
Hoyer lift	€443.40	0.0	one-time
Hospital bed	€7,423.86	0.0	1.0
Speech devices	€1,089.00	0.0	one-time
Car/home modification	€3,489.00	0.0	one-time
N° of physiotherapist sessions	€35.00	170.8	169.9

Table 1. Resource utilization by ambulatory and non-ambulatory patients. Estimated based off expert ascribed typical resource use

Results

- Among non-ambulatory patients, 56% were estimated to have difficulties (score 3-4 in FA-ADL) with dressing/hygiene/feeding, 29% with swallowing/speech, 25.5% with bladder function, and 37.3% with sitting posture.
- Additionally, 45% had cardiomyopathy and/or scoliosis.
- Non-ambulatory patients had an estimated 25.8 specialist visits and 273 days of home health nursing, along with continuous caregiving per annum compared to 8.1 specialist visits and 30 days of home health nursing for ambulatory patients.
- Assistive Support Instruments were used continuously, physiotherapy sessions remained constant at 170 per year.
- From the NHS perspective, the estimated annual cost for non-ambulatory patients was €39,843 with also additional one-time costs of €23,428 due to loss of ambulation compared to €8,545 annually for ambulatory patients (Figure 1).
- When considering the societal perspective, the estimated annual costs per non-ambulatory patient was €99,354 plus €26,917 one-time costs vs €24,507 annually per ambulatory patient (Figure 1).

Ambulatory	Non-Ambulatory
National Healthcare Service	
Annual cost	Annual cost
€ 8,545	€ 39,843
One-time Cost	One-time Cost
€ 0.00	€ 23,428
Society	
Annual cost	Annual cost
€ 24,507	€ 99,354
One-time Cost	One-time Cost
€ 0.00	€ 26,917

Figure 1. Estimated National Healthcare Service and Society annual and one-time by ambulatory (green) and non-ambulatory (blue) patients. Estimated based off expert ascribed typical resource use

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