

Patient Journeys are **info-graphical overviews** that visualize patients' needs in the care of their rare disease.

Because Patient Journeys are designed from the **patient's perspective**, they allow clinicians to **effectively address the needs** of rare disease patients.

Find a detailed version of this patient journey on our website.

PATIENT JOURNEY FRIEDREICH'S ATAXIA (FA)			
PROCESSES	1. First symptoms	2. Diagnosis	3. Treatment
Process	Initially, walking is the main, unproblematic activity. As the disease progresses, patients experience difficulties with balance and coordination, leading to frequent falls and gait abnormalities. The first signs of the disease are often subtle and may be mistaken for other conditions.	Diagnosis is typically made through a combination of clinical history, physical examination, and genetic testing. The process involves a series of consultations with different specialists, including neurologists, geneticists, and sometimes psychologists.	Treatment is primarily symptomatic and focuses on managing the symptoms of the disease. This includes physical therapy, occupational therapy, and speech therapy. There is no cure for the disease, and treatment aims to improve quality of life and slow down the progression of the disease.
Challenges	Patients with Friedreich's Ataxia face several challenges, including difficulties with walking, balance, and coordination. They may also experience cognitive and emotional challenges, such as anxiety and depression. The disease is often progressive, leading to a decline in physical and cognitive abilities over time.	Diagnosis can be challenging due to the rarity of the disease and the need for specialized testing. Patients may face delays in diagnosis and may need to travel to specialized centers for testing and treatment.	Treatment is often limited and may not fully address the needs of the patient. Patients may face challenges with accessing specialized services and may need to travel to specialized centers for treatment.
Goals	Patients and their families may have several goals, including improving walking and balance, managing symptoms, and maintaining quality of life. They may also want to learn more about the disease and its progression.	Patients and their families may have several goals, including getting a diagnosis, understanding the disease, and accessing specialized services. They may also want to participate in research and advocacy efforts.	Patients and their families may have several goals, including managing symptoms, improving quality of life, and slowing down the progression of the disease. They may also want to participate in research and advocacy efforts.



PATIENT JOURNEY

Friedreich's Ataxia (FA)

different needs
at different times



Was this patient journey helpful?
Help us improve patient care
and participate in our short survey!



European Reference Network
for Rare Neurological Diseases
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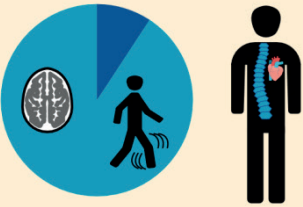





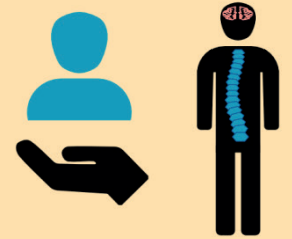
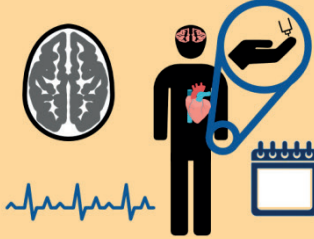








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**European
Reference
Network**
for rare or low prevalence
complex diseases
Network
Neurological Diseases
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**Friedreich's
Ataxia
Research
Alliance
IRELAND**

	First Symptoms	Diagnosis	Treatment	Monitoring
Disease	<p>91% neurological symptoms. 9% non-neurological symptoms: scoliosis and heart trouble.</p> 	<p>Genetic testing is available, but gene changes in FA are not recognized using standard NGS.</p> 	<p>One disease-specific therapy in Europe and USA approved, several clinical trials ongoing.</p> 	<p>Referral to expert center. Multidisciplinary team for heart condition and diabetes.</p> 
Clinic	<p>Assessment of symptoms and referral to relevant specialists.</p> 	<p>Genetic counselling for parents regarding future pregnancies and siblings above 18.</p> 	<p>Mental health support, neurological and scoliosis assessment.</p> 	<p>Annual review of mobility, ability to do daily activities, heart, diabetic risk etc.</p> 
Challenges	<p>Confusion and complexity of symptoms leads to frequent misdiagnosis.</p> 	<p>Consider diagnosis in all age groups, since 1% of all presenta- tions are in those 60+ years.</p> 	<p>Maintaining personal autonomy and ability to walk, access to currently available treatments.</p> 	<p>Children may isolate themselves. Parents are often unsure how to treat their child with FA.</p> 
Goals	<p>Take patients with multi-system complaints seriously, i.e. clumsiness, fatigue, back pain.</p> 	<p>Genetic counselling and testing for extended family, including cousins, to inform about genetic risk factors.</p> 	<p>Care guidelines should be shared with person with FA. Worldwide access to therapy.</p> 	<p>Maximize the potential to live a life as normal as possible, e.g. driving and part-time work.</p> 

FA
NGS

Friedrich's Ataxia
Next Generation Sequencing
(mapping of entire genome)

Please note that specific terms (e.g. home care services, general physician, physiotherapy) do not include the same services in all EU countries and might differ from country to country. Patient advocacy groups can often provide support and resources for patients and families.

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