

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)			
PHASE	1 - First symptoms	2 - Diagnosis	3 - Treatment
Disease	<p>Difficult walking in the dark, unsteadiness in standing or walking, clumsiness, frequent falls and/or loss of consciousness.</p> <p>Loss of speech control with poor balance or falls.</p> <p>Onset of non-neurological symptoms (e.g. diabetes or heart disease) may precede neurological symptoms.</p>	<p>Genetic testing for FA can be done even if there is a clinical suspicion of another rare genetic condition (MPS) and requires specific tests.</p>	<p>One disease specific therapy approved in Europe and USA - vitamin E, which is effective in slowing down the progression of cerebellar ataxia.</p> <p>Other drugs may be indicated when the cerebellum is severely affected.</p> <p>• Cerebellar ataxia can be treated with medication.</p>
Obstacles	<p>Assessment of symptoms and when to request genetic testing.</p> <p>Medication issues that include monitoring, effectiveness, side effects, availability and other treatment options depending on need.</p>	<p>Counseling</p> <ul style="list-style-type: none"> • genetic testing for the disease • testing options (genotype and allele frequency) • allele frequency testing • carrier status • family history • genetic testing of reproductive age 	<p>Psychological and social support for the individual and all family members for the diagnosis and treatment.</p> <ul style="list-style-type: none"> • Support of the individual and family members • Support of the individual and family members • Support of the individual and family members • Support of the individual and family members
Challenges	<p>How to confirm the diagnosis of FA with a genetic test.</p> <p>Diagnosis of FA can be difficult and may not be apparent in the laboratory or test results at all times.</p> <p>How to have genetic testing.</p> <p>The allele frequency test is not available in all laboratories or in all countries.</p>	<p>Access to comprehensive care for FA is not available in all countries.</p> <p>Encourage</p> <ul style="list-style-type: none"> • use of genetic testing in the individual and family members • genetic testing in the individual and family members • genetic testing in the individual and family members • genetic testing in the individual and family members 	<p>Patients should be supported and advised about how to communicate the diagnosis of FA to other family members.</p> <p>The individual may not be able to communicate with other family members and may need support.</p> <p>As a result, the genetic test should be performed and should be available to the patient with FA.</p>
Goals	<p>Each patient consistently receives in disease diagnosis and treatment.</p> <p>Each patient consistently receives in disease diagnosis and treatment.</p> <p>Each patient consistently receives in disease diagnosis and treatment.</p>	<p>Access to comprehensive care for FA is not available in all countries.</p> <p>Encourage</p> <ul style="list-style-type: none"> • use of genetic testing in the individual and family members • genetic testing in the individual and family members • genetic testing in the individual and family members • genetic testing in the individual and family members 	<p>Patients should be supported and advised about how to communicate the diagnosis of FA to other family members.</p> <p>The individual may not be able to communicate with other family members and may need support.</p> <p>As a result, the genetic test should be performed and should be available to the patient with FA.</p>



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!



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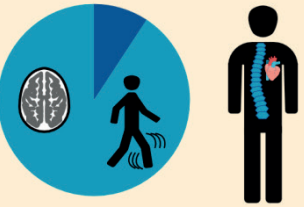





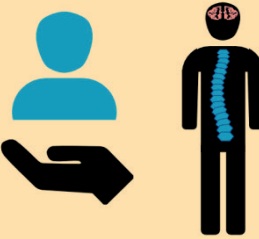
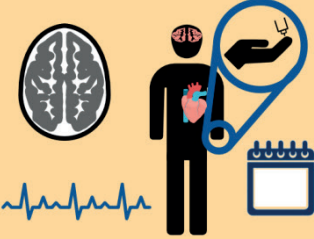





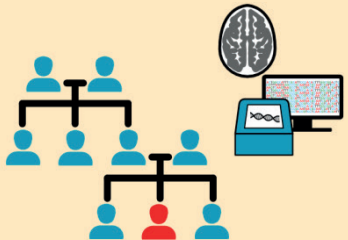




European Reference Network
for rare or low prevalence complex diseases

Network Neurological Diseases (ERN-RND)



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, as 1% of those with FA are over 60 years old.</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 to avoid FA presenting in cousins</p> 	<p>Care guidelines should be shared with person with FA. Worldwide access to therapy</p> 	<p>Maximize the potential to live as normal a life as possible, e.g. driving and part-time work.</p> 

FA
NGS
Friedreich'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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Neurological Diseases
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