PATIENT JOURNEY

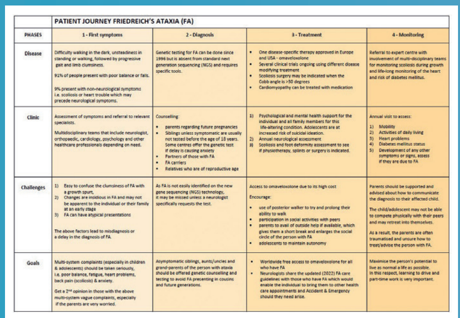
Friedreich's Ataxia (FA)

different needs at different times

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.



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

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for Rare Neurological Diseases
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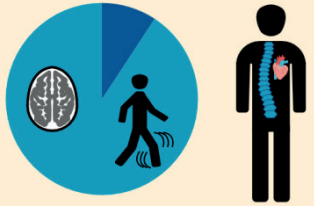
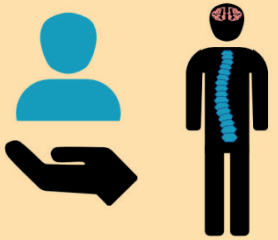
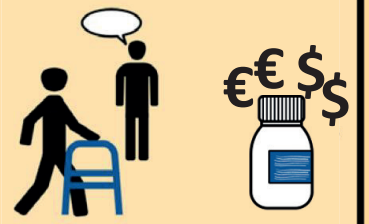
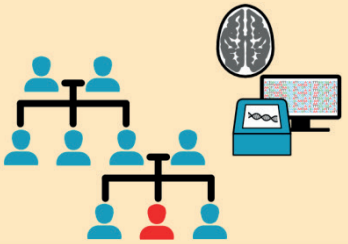
European Reference Network
for rare or low prevalence complex diseases
Network
Neurological Diseases
(ERN-RND)

Friedreich's Ataxia Research Alliance IRELAND

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

FA Friedreich's Ataxia
NGS 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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