PATIENT JOURNEY

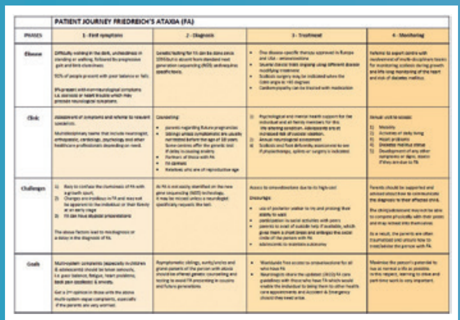
Friedreich's Ataxia (FA)

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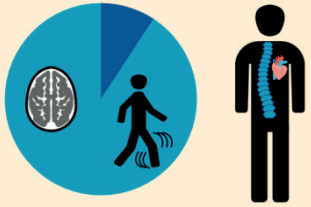
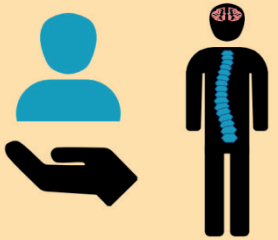
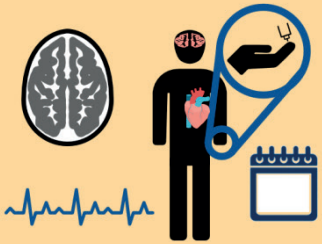
European Reference Network
for rare or low prevalence complex diseases
Network
Neurological Diseases
(ERN-RND)

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, since 1% of all presentations are in those 60+ years. 	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, including cousins, to inform about genetic risk factors. 	Care guidelines should be shared with person with FA. Worldwide access to therapy. 	Maximize the potential to live a life as normal 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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