

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.

For more information, please visit:

<https://www.ern-rnd.eu/patient-journey-friedreichs-ataxia/>



Friedreich's Ataxia

The patient journey



A visual description of what patients need and how clinicians can address them







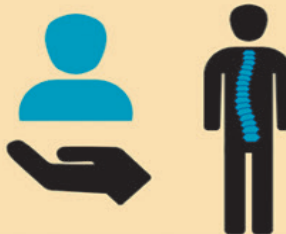






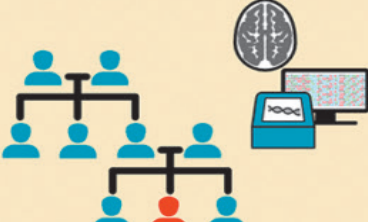

FARA | Friedrich's
Ataxia
Research
Alliance



European
Reference
Network

for rare or low prevalence
complex diseases

 **Network**
Neurological Diseases
(ERN-RND)

	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>Several clinical trials ongoing, but no effective disease- modifying therapies yet</p> 	<p>Referral to expert centre. Multi- disciplinary 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 young adults</p> 	<p>Mental health support and scoliosis assessment</p> 	<p>Annual neurological and cardiac assessment, diabetes check-up, review mobility and daily living</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>Enlarge social circle, maintain personal autonomy, prolong walking ability with use of aids</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 are available and should be shared with person with FA</p> 	<p>Maximize the potential to live as normal a life as possible, e.g. driving and part-time work.</p> 