

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)

Disease

First Symptoms

91% neurological symptoms.
9% non-neurological symptoms:
scoliosis and heart trouble

Diagnosis

Genetic testing is available, but
gene changes in FA are not
recognized using standard NGS

Treatment

One disease-specific therapy in
Europe and USA approved,
several clinical trials ongoing

Monitoring

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
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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This work is generated within the European Reference Network for Rare Neurological Diseases

