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 a digital version of the patient journey scan the QR code or visit the ERN-RND website:

www.ern-rnd.eu/patient-journey-friedreichs-ataxia/

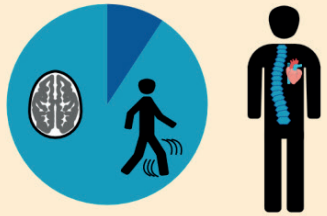
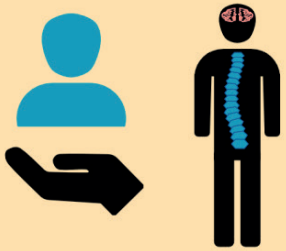
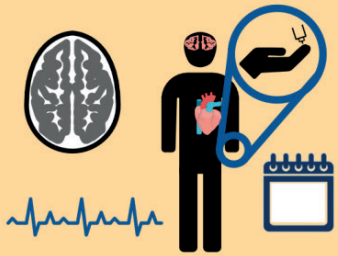
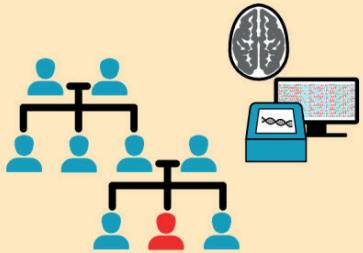
# Friedreich's Ataxia

## The patient journey

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

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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