

The 'Patient Journey' for Rare Neurological Diseases – Why, how and for who?



ePAG

European Patient Advocacy Group

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Introduction

The patient journey is a series of interactions and experiences a patient has with the healthcare system from initial symptoms to treatment, and follow-up. To achieve a patient journey, patient representatives usually complete a mapping exercise of the needs of each rare syndrome across the different stages. The final Patient Journey connects professional expert guidelines, with foreseen medical interventions (when indicated) screening, treatment (when available) with patient needs-both medical and psychological. The patient journey is reviewed by both patient and professional experts before publication. It is an ideal way to harness all available knowledge on a rare disease.

Why?

Within ERN-RND after developing disease-specific patient journeys for Friedreich's ataxia, Huntington's disease, Cervical Dystonia, Multiple System Atrophy and Hereditary Spastic Paraplegia, it became clear that many experiences and challenges are shared across conditions. As a result, it was essential to consolidate these findings into a [Patient Journey for Rare Neurological Diseases](#).



Methodology – How?

- Meeting face-to-face in Stockholm in May 2023
- Focus group with clinicians in February 2024 to gather their perspective
- Online meetings: consensual review of each section
- Text version turned into graphical version by Katerina
- Validation with clinicians in July 2025



Results

	Pre-manifest	First symptoms	Diagnosis	Treatment	Monitoring
Disease	<p>Rare neurological diseases (RND) often cause great anxiety within a family.</p>	<p>Many RND share common multiple symptoms - usually progressive, some episodic.</p>	<p>Misdiagnosis, delayed diagnosis, no diagnosis in some cases. Incomplete genetic diagnosis.</p>	<p>Very few disease modifying treatments; Only observation & symptom management to prevent avoidable disability.</p>	<p>Disease progression differs from patient to patient, so it is difficult to predict at the first consultation.</p>
Clinic	<p>No signs. Only genetic testing can prove it.</p>	<p>Progressive, some degenerative rare diseases.</p>	<p>Communicate risk to the extended family. Family planning for parents as applicable. Genetic counselling.</p>	<p>Consistent and nurtured doctor/patient relationship in a multi-disciplinary clinic.</p>	<p>Encourage healthy lifestyle, mental health review, carer support, access to patient organizations.</p>
Challenges	<p>Fear of developing progressive RND. Value of testing vs. not testing.</p>	<p>Lack of knowledge among medical profession to recognise first symptoms.</p>	<p>Finding the path to the right expertise for correct diagnosis, genetic counselling delayed, stigma about inherited disease.</p>	<p>Lack of treatment, knowledge and access to clinical trials. Financial burden for those affected.</p>	<p>Knowledge of care guidelines, prediction of progression difficult, lack of palliative support.</p>
Goals/Needs	<p>Consider patient preferences during genetic counselling.</p>	<p>Consult ERN-RND guidelines. Faster access to genetic testing. Improve access to expertise.</p>	<p>Timely genetic diagnosis and counselling. Genetic testing for RND families, pre-natal assessment in RND families.</p>	<p>Multidisciplinary, integrated & holistic care. Consider family perspective. Access to timely treatment.</p>	<p>Improved information sharing and awareness of RND by all stakeholders.</p>

For Who?

The common rare neurological Patient Journey is designed to support both clinicians, in their interactions with patients, and patients themselves, helping them better understand their own path. It highlights recurring gaps in care & identifies key areas for improvement. It is important to remember that there is a considerable heterogeneity between healthcare services in the many European countries.

Raising awareness Raise awareness and understanding among general clinicians, medical/neurology students & general practitioners

Empowering patients Empower patients to communicate pro-actively with healthcare professionals (expectations and treatment dissatisfaction)
Guide patients to relevant online sources and enable them to understand their symptoms

Remind them the importance of mental health support for disease which are life altering and most often progressive

Facilitate patients' social contact with patient organisations and other affected persons which will help to motivate them to maintain social relationships

Empowering healthcare professionals Help healthcare professionals know the value of referring their patients to patients associations for information and social interaction

Support a multidisciplinary treatment approach throughout all phases of the patient journey while recognising individual patients' information needs and explore all possible treatment options

Requests to policy makers Promote the recognition and reimbursement of treatments, when available, across all European countries, including support for cross-border healthcare access

Given the lack of knowledge about rare disease and limited resources for education, continue to fund the ERN-RND, and consider using telemedicine options, digital solutions to provide training to local healthcare providers



European Reference Network

for rare or low prevalence complex diseases

Network Neurological Diseases (ERN-RND)

Funded by the European Union