Patient Journey for Rare Neurological Diseases - Detailed Version

Please be aware that not all RND are genetic. Text in italic denotes those conditions where the information does not apply, e.g. most forms of Frontotemporal Dementia, non-genetic Dystonia, Chorea, Atypical Parkinsonism

PHASES	Pre-manifest	First Symptoms	Diagnosis	Treatment	Monitoring
Disease	Rare neurological diseases (RND) often cause great anxiety within a family.	Many RND share common multiple symptoms which are usually slowly progressive but occasionally are episodic (episodic ataxia).	• Misdiagnosis, delayed diagnosis, no diagnosis in some cases • *Incomplete genetic diagnosis*	• Few, if any disease-modifying treatments are available. • Observation and symptom management to improve the quality of life	Disease progression differs from patient to patient so it is difficult to predict at the first consultation.
Clinic	No signs. *Some asymptomatic genetic diseases can be diagnosed by genetic testing when available.* Other RND, e.g. most forms of Frontotemporal Dementia and Multiple System Atrophy are non-genetic.	• First symptoms may be unspecific and might make the diagnosis difficult • Some RND are degenerative in nature which leads to progression in the individuals' symptoms over time • Lack of knowledge	When diagnosis of a genetic disease is made, it is important to • communicate risk to the extended family • advise in regards to family planning for parents as applicable *Genetic counselling is preferable, but not always immediately available.*	• Consistent and nurtured doctor/patient relationship in a multi-disciplinary clinic • Timely access to treatment if available, and/or clinical trial access for RND	• Regular assessment of RND in keeping with European or global guidelines • Mental health review of patient • Carer support • Advise a healthly lifestyle, including exercise as appropriate • HCPs'should recommend RND individuals to get support from an expert patient organisation
Challenges	• Fear of developing progressive RND • *Value of genetic testing vs. not testing*	Lack of awareness among non-specialist professionals to recognise first symptoms ~~and~~ causes delay in diagnosis	• Waiting for a diagnosis • People not investigated in a systematic way • *Genetic counselling not readily available when a diagnosis is made* • Stigma associated with inherited disease • For diseases with a young onset several societal, familiar and life course challenges may arise	• Lack of treatment • Lack of knowledge within local services about RND and subsequent lack of access to clinical trials • Financial burden for the RND family • Self-directed care is difficult as one navigates between the different systems of care	• Medical professional caring for the RND patient not aware of care guidelines for the specific RND • RND progression is difficult to predict • Increased financial and emotional burden as the disease progresses • Inadequate support and care for those who have a terminal illness
Goals/Needs	• *Consider patient preferences in consultation with a geneticist and psychiatrist (if possible) on predictive genetic testing* • *Best practice currently suggests that individuals be over 18 years before they are tested if they have no symptoms of the genetic disease, which the family have except in rare circumstances – where there is a life-saving treatment*	• People with neurological disease should be investigated in line with diagnostic guidelines which are available on the ERN-RND website • *Faster access to genetic testing for the RND family* • Access to appropriate specialised investigations as needed	• *Genetic diagnosis in a timely manner where possible* • *Genetic testing for RND families where applicable* • Pre-natal assessment in RND families who have an identifiable autosomal dominant disease (when available and allowed in your country)	• Multidisciplinary, integrated care & holistic care for rare disease with central accessible RND resource for local HCPs • Consider family perspective and support their needs where possible • Access to timely treatment to minimize costs and disease burden for society • Scientists to get input from those with RND regarding clinical trial outcomes; i.e. is ability to communicate more important than ability to walk • Learn how to cope with the disease (both the patient and her/his family)	• Collaborative communication between the patient and the clinicians about the effect of the medical treatment • Support to care for the person with RND optimally with improved access to equipment and resources • Patient experts via patient organisations can help educate those with a RND

Please note that specific terms (e.g. home care services, general physician, physiotherapy) may not include the same services dependent on the individual EU country. Patient advocacy groups can often provide support and resources for patients and families.

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Network Neurological Diseases (ERN-RND)