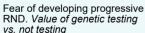
## Patient Journey for Rare Neurological Diseases - Graphical Version

**Pre-manifest First Symptoms Diagnosis Treatment Monitoring** Disease Very few diseases with modifying Rare neurological diseases (RND) Many RND share common multiple Misdiagnosis, delayed diagnosis, treatments; observation & Disease progression differs from often cause great anxiety within no diagnosis in some cases. symptoms - usually progressive, patient to patient, so it is difficult symptom management to improve a family. some episodic. Incomplete genetic diagnosis. to predict at the first consultation. the quality of life. No signs. Genetic diseases can be diagnosed at the asympto-First symptoms may be Communicate risk to the extended Consistent and nurtured doctor/ Encourage healthy lifestyle, menmatic stage by genetic testing (if tal health review, carer support. unspecific and might family. Family planning for parents patient relationship in a multiavailable), and receive support. access to patient organisations. make the diagnosis difficult. as applicable. Genetic counselling disciplinary clinic. Challenges



**Goals/Needs** 





Lack of awareness among non-specialist professionals to recognise first symptoms.



Finding the path to the right expert for correct diagnosis, genetic counselling delayed. stigma about inherited disease.



Lack of treatment, knowledge and access to clinical trials. Financial burden for those affected.



Knowledge of care guidelines; Prediction of progression difficult: Lack of palliative support.





for rare or low prevalence complex diseases

Network Neurological Diseases (ERN-RND)

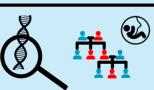




Consider patient preferences during genetic counselling. Asymptomatic people >18 years.



Consult ERN-RND guidelines; Faster access to genetic testing; Improve access to expertise.



Timely genetic diagnosis and counselling. Genetic testing for RND families, pre-natal assessment in RND families.



Multidisciplinary, integrated & holistic care. Consider family perspective. Access to timely treatment.



Improved information sharing and awareness of RND by all stakeholders.

Please be aware that not all rare neurological diseases 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.

Please note that specific terms

(e.g. home care services, general physician, physiotherapy) may not include the same services dependnet on the individual EU country. Patient advocacy groups

can often provide support and resources for patients and families.

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