Diagnostic flowchart for Hereditary Spastic Paraplegias (HSP)

suggestions for comprehensive phenotyping in HSP

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Introduction to the European Reference Network for Rare Neurological Diseases (ERN-RND):

ERN-RND is a European Reference Network established and approved by the European Union. ERN-RND is a healthcare infrastructure which focuses on rare neurological diseases (RND). The three main pillars of ERN-RND are (i) network of experts and expertise centres, (ii) generation, pooling and dissemination of RND knowledge, and (iii) implementation of e-health to allow the expertise to travel instead of patients and families.

ERN-RND unites 32 of Europe’s leading expert centres in 13 Member States and includes highly active patient organizations. Centres are located in Belgium, Bulgaria, Czech Republic, France, Germany, Hungary, Italy, Lithuania, Netherlands, Poland, Slovenia, Spain and the UK.

The following disease groups are covered by ERN-RND:

- Ataxias and Hereditary Spastic Paraplegias
- Atypical Parkinsonism and genetic Parkinson’s disease
- Dystonia, Paroxysmal Disorder and Neurodegeneration with Brain Ion Accumulation
- Frontotemporal Dementia
- Huntington’s Disease and other Chorea
- Leukodystrophies

Specific information about the network, the expert centres and the diseases covered can be found at the networks web site www.ern-rnd.eu.

Recommendation for clinical use:

The European Reference Network for Rare Neurological Diseases developed the Diagnostic Flowchart for HSP to help guide the diagnosis. The Reference Network recommends the use of this Diagnostic Flowchart.
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METHODOLOGY

The development of the Diagnostic Flowchart was done by the Disease group for Ataxia and Hereditary Spastic Paraplegias of ERN-RND.

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Flowchart development process:

- Development of flowchart – June – November 2017
- Discussion/Revision in ERN-RND disease group – November 2017 – June 2018
- Consent on diagnostic flowchart during ERN-RND annual meeting 2018 – 08/06/2018
- Consent on document by whole disease group – 15/11/2018
HSPs are phenotypically highly variable and genetically highly heterogeneous. Single gene testing other than indicated below is therefore not recommended. Instead, comprehensive NGS-based gene panels or whole exome/genome sequencing should be employed as a first line diagnostic testing.