

Diagnostic flowchart for adult ataxias

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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
- Huntingtons' Disease and other Choreas
- 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 adult ataxias 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.

Disease group for Ataxia and Hereditary Spastic Paraplegias:

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Patient representatives:

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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:30 November 2018
- Consent on document by whole disease group 05/02/2019



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Diagnostic flowcharts -Ataxias

Exclusion of acquired causes*** in case of negative family history, (sub)acute onset, specific medical history, etc.

***Common Acquired Causes: auto immune diseases (MS, sarcoidosis, celiac disease, etc), toxic reaction, head trauma, cerebral palsy, tumor, stroke, infections, vitamine deficiency, paraneoplastic syndromes

CHECK for presence/absence: (1) peripheral neuropathy-sensory neuronopathy; (2) Cerebellar /brainstem/cerebral MRI findings

