



# Diagnostic flowcharts for Dystonia: (1) In adults (2) In children & adolescents

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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 Parkinsons' Disease
- Dystonia, Paroxysmal Disorder and Neurodegeneration with Brain Ion Accumulation
- Frontotemporal Dementia
- Huntingtons' 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](http://www.ern-rnd.eu).

## Recommendation for clinical use:

**The European Reference Network for Rare Neurological Diseases developed the Diagnostic Flowcharts for Dystonia to help guide the diagnosis of Dystonia patients. The Reference Network recommends the use of these Diagnostic Flowcharts.**



#### Disclaimer:

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## **METHODOLOGY**

The development of the Diagnostic Flowcharts for Dystonia was done by the Disease group for Dystonia, Paroxysmal Disorder and NBIA of ERN-RND.

### Disease group for Dystonia, Paroxysmal Disorder and NBIA:

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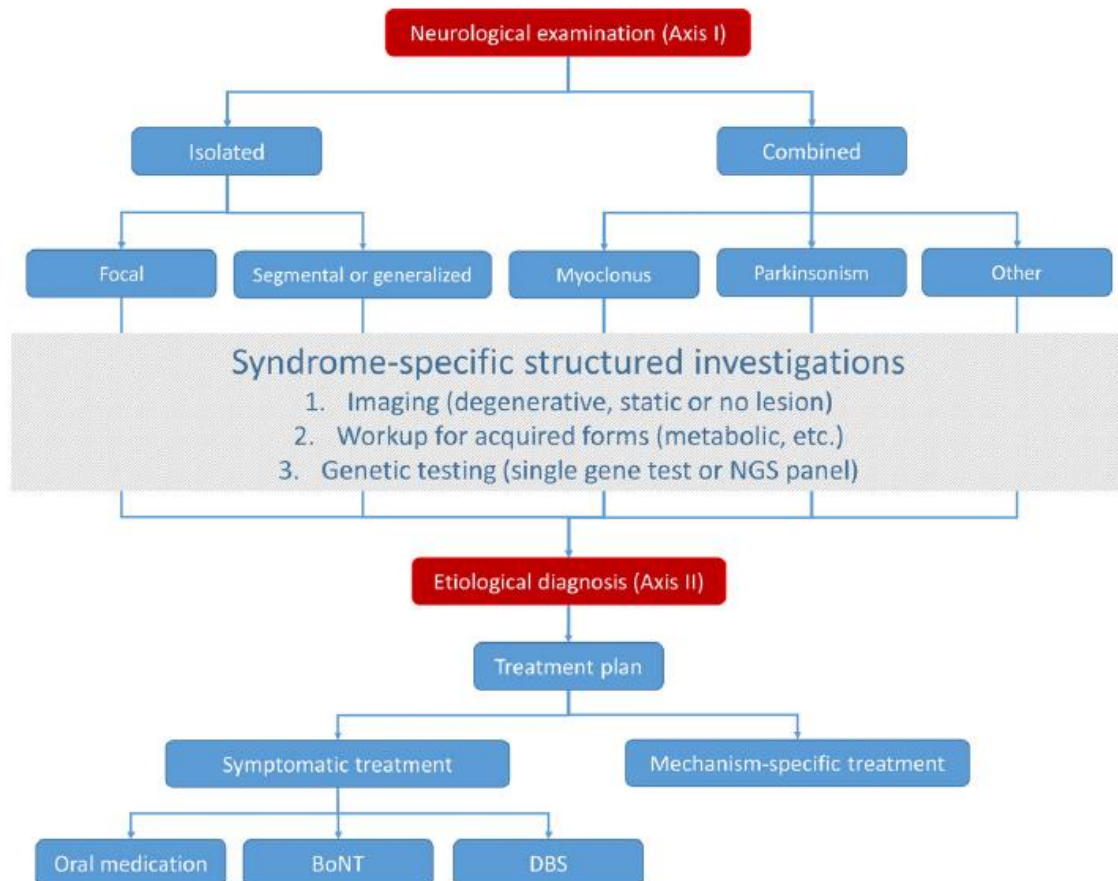
## Flowchart development process:

- Development of flowcharts – June – November 2017
- Discussion/Revision in ERN-RND disease group– November 2017 – June 2018
- Consent on diagnostic flowcharts during ERN-RND annual meeting 2018 – 08/06/2018
- Consent on document by whole disease group – 26/09/2018



## **Diagnostic flowchart for Dystonia in adults**

(according to A. Albanese, M. Di Giovanni and S. Lalli: Dystonia: Diagnosis and Management, Eur J Neurol. 2018 Jul 23. doi: 10.1111/ene.13762. [Epub ahead of print])



**Figure 2** Clinical strategy from examination to treatment plan. Following examination, phenomenology guides diagnostic testing. The information collected allows a treatment plan, whether symptomatic or mechanism specific, to be defined. A listing of specific disease-modifying treatments for dystonia syndromes has recently been compiled [94]. BoNT, botulinum neurotoxin; DBS, deep brain stimulation; NGS, next-generation sequencing. [Colour figure can be viewed at [wileyonlinelibrary.com](http://wileyonlinelibrary.com)]

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## Diagnostic flowchart for Dystonia in children and adolescents

(according to van Egmond ME, Kuiper A, Eggink H, et al. J Neurol Dystonia in children and adolescents: a systematic review and a new diagnostic algorithm Neurosurg Psychiatry 2015;86:774–781.)

Full article can be found here: <http://dx.doi.org/10.1136/jnnp-2014-309106>

Diagnostic algorithm of dystonia in children and adolescents

