European Reference Networks

European Reference Network for rare or low prevalence complex diseases

Network: Neurological Diseases (ERN-RND)

Coordinator: Universitätsklinikum Tübingen — Deutschland

TREATMENT ALGORITHM FOR FOCAL AND GENERALIZED DYSTONIAS

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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 64 of Europe's leading expert centres as well as 4 affiliated partners in 24 member states and includes highly active patient organizations. Centres are located in Austria, Belgium, Bulgaria, Croatia, Cyprus, Czech Republic, Denmark, Estonia, Finland, France, Germany, Greece, Hungary, Ireland, Italy, Latvia, Lithuania, Luxembourg, Malta, Netherlands, Poland, Slovenia, Spain and Sweden.

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 Iron Accumulation
- Frontotemporal Dementia
- Huntington's Disease and other Chorea
- Leukodystrophies

*Specific information about the network, the expert centers and the covered diseases can be found on the network's website **www.ern-rnd.eu**.*

Recommendation for clinical use:
The European Reference Network for Rare Neurological Diseases strongly recommends the use the following treatment algorithm for focal and generalised dystonia.

DISCLAIMER

Clinical practice guidelines, practice advisories, systematic reviews and other guidance published, endorsed or affirmed by ERN-RND are assessments of current scientific and clinical information provided as an educational service.

The information (1) should not be considered inclusive of all proper treatments, methods of care, or as a statement of the standard of care; (2) is not continually updated and may not reflect the most recent evidence (new information may emerge between the time information is developed and when it is published or read); (3) addresses only the question(s) specifically identified; (4) does not mandate any particular course of medical care; and (5) is not intended to substitute for the independent professional judgement of the treating provider, as the information does not account for individual variation among patients. In all cases, the selected course of action should be considered by the treating provider in the context of treating the individual patient. Use of the information is voluntary. ERN-RND provided this information on an "as is" basis, and makes no warranty, expressed or implied, regarding the information. ERN-RND specifically disclaims any warranties of merchantability or fitness for a particular use or purpose. ERN-RND assumes no responsibility for any injury or damage to persons or property arising out of or related to any use of this information or for any errors or omissions.

METHODOLOGY

The endorsement of the treatment algorithm for focal and generalized dystonia was done by the Disease group for Dystonia, Paroxysmal Disorder and NBIA of ERN-RND.

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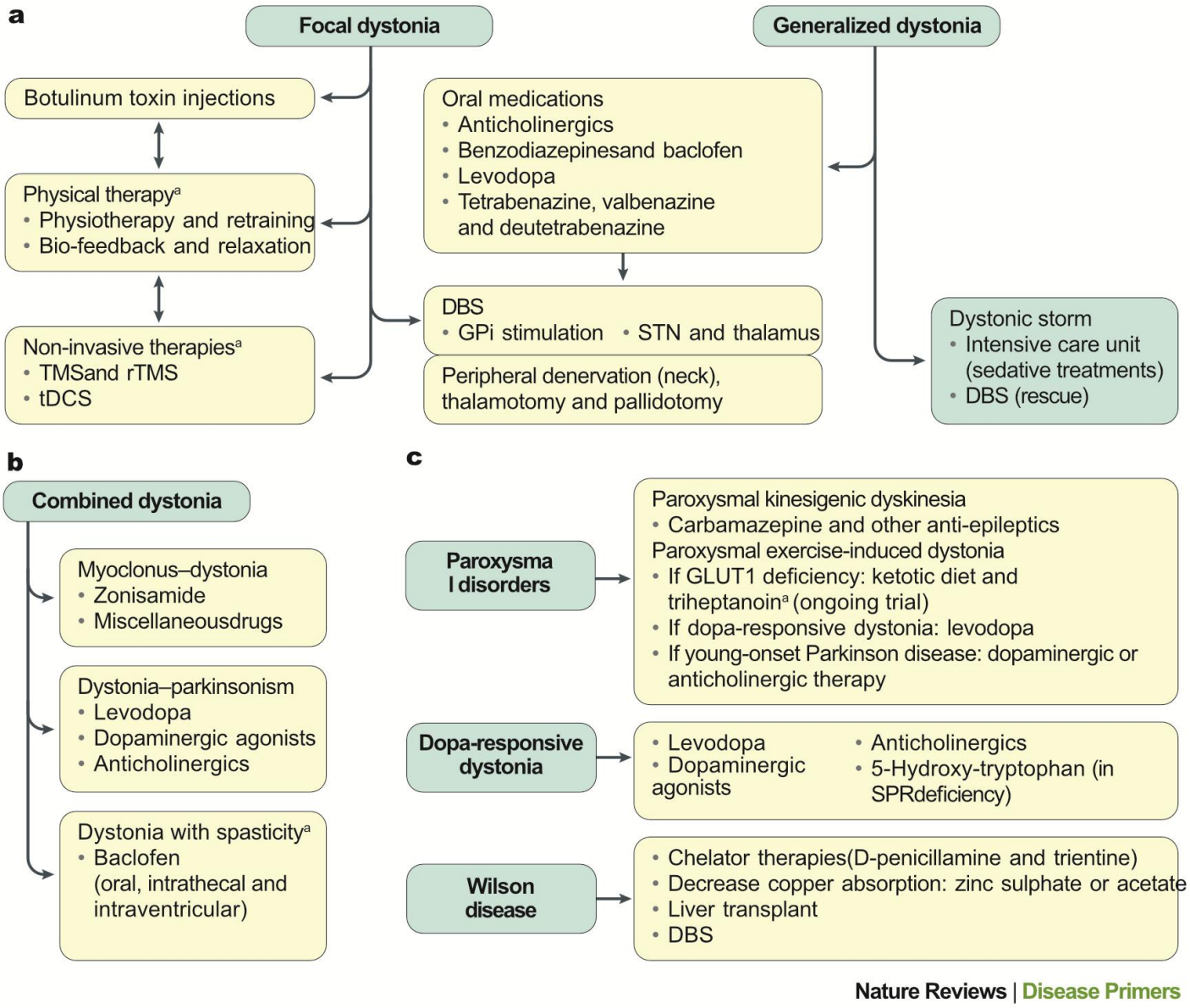
Endorsement process:

- Consent to endorse document by whole disease group – 18 June 2019
- Last revision and consent on updates by whole disease group – 11 October 2024
- Update published – 04 June 2025

REFERENCE

Balint, B., Mencacci, N.E., Valente, E.M. et al. Dystonia. Nat Rev Dis Primers 4, 25 (2018). https://doi.org/10.1038/s41572-018-0023-6

TREATMENT ALGORITHM



Treatment algorithm for dystonia. The treatment of dystonia remains symptomatic, and the therapeutic approach is largely orientated at the distribution of dystonic symptoms (for example, whether dystonia is focal or generalized (part **a**)). Therapy comprises botulinum toxin injections (for focal or segmental dystonia), oral medications (mainly for generalized dystonia) and deep brain stimulation (DBS) (for generalized, focal or segmental dystonia), which can be complemented by physical therapies and non-invasive stimulation techniques, although the efficacy of the latter therapy is still to be validated. Treatment of combined dystonia takes into account the other presenting signs (part **b**). Specific dystonia syndromes have specific treatment approaches, such as paroxysmal disorders, dopa-responsive dystonia and Wilson disease (part **c**). A comprehensive discussion of specific treatment approaches in hereditary (mostly combined) dystonias can be found elsewhere168. GLUT1, glucose transporter type 1, erythrocyte/brain; GPi, globus pallidus internus; rTMS, repetitive transcranial magnetic stimulation; SPR, sepiapterin reductase; STN, subthalamic nucleus; tDCS, transcranial direct-current stimulation; TMS, transcranial magnetic stimulation. [a]The efficacy of these treatments is still to be validated.

Direct access to original article via https://www.nature.com/articles/s41572-018-0023-6, see also reference

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