European Reference Networks

European Reference Network for rare or low prevalence complex diseases

Network: Neurological Diseases (ERN-RND)

Coordinator: Universitätsklinikum Tübingen — Deutschland

MANAGEMENT OF NON-PKAN GENETIC DEFECTS

ERN-RND-endorsed review articles

Endorsed by ERN-RND: 16th June 2020

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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 (RNDs). 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 Disorders and Neurodegeneration with Brain Iron Accumulation
- Frontotemporal Dementia
- Huntingtons' Disease and other Choreas
- Leukodystrophies

Specific information about the network, the expert centres and the diseases covered can be found on the network's website

www.ern-rnd.eu.

Recommendation for clinical use:
Currently, there are no guidelines available for the management of non-PKAN genetic defects.
The European Reference Network for Rare Neurological Diseases strongly recommends following the guidance given in the review articles below for the management of non-PKAN genetic defects.

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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 review articles for non-PKAN genetic defects was done by the Disease group for Dystonia, Paroxysmal Disorders and NBIA of ERN-RND.

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Endorsement process:

- Consent to endorse documents by whole disease group – 16.06.2020

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- **Beta-Propeller Protein-Associated Neurodegeneration**. Allison Gregory, Manju A Kurian, Tobias Haack, Susan J Hayflick, Penelope Hogarth, Margaret P Adam, Holly H Ardinger, Roberta A Pagon, Stephanie E Wallace, Lora JH Bean, Karen Stephens, Anne Amemiya, editors. In: GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2020. 2017 Feb 16.

Due to the regulations on the protection of intellectual property, we are not allowed to print the actual therapeutic algorithm in this document.

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https://ec.europa.eu/health/ern_en

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