European Reference Networks

European Reference Network for rare or low prevalence complex diseases

Network: Neurological Diseases (ERN-RND)

Coordinator: Universitätsklinikum Tübingen – Deutschland

DIAGNOSTIC FLOWCHART FOR PAROXYSMAL DISORDERS

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW PREVALENCE AND COMPLEX DISEASES

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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 (NBIA)
- Frontotemporal Dementia
- Huntington's Disease and other Choreas
- Leukoencephalopathies

*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 developed the Diagnostic Flowchars for Paroxysmal Disorders to help guide the diagnosis of Paroxysmal Disorder patients. The Reference Network recommends the use of this Diagnostic Flowchart.

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.

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METHODOLOGY

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

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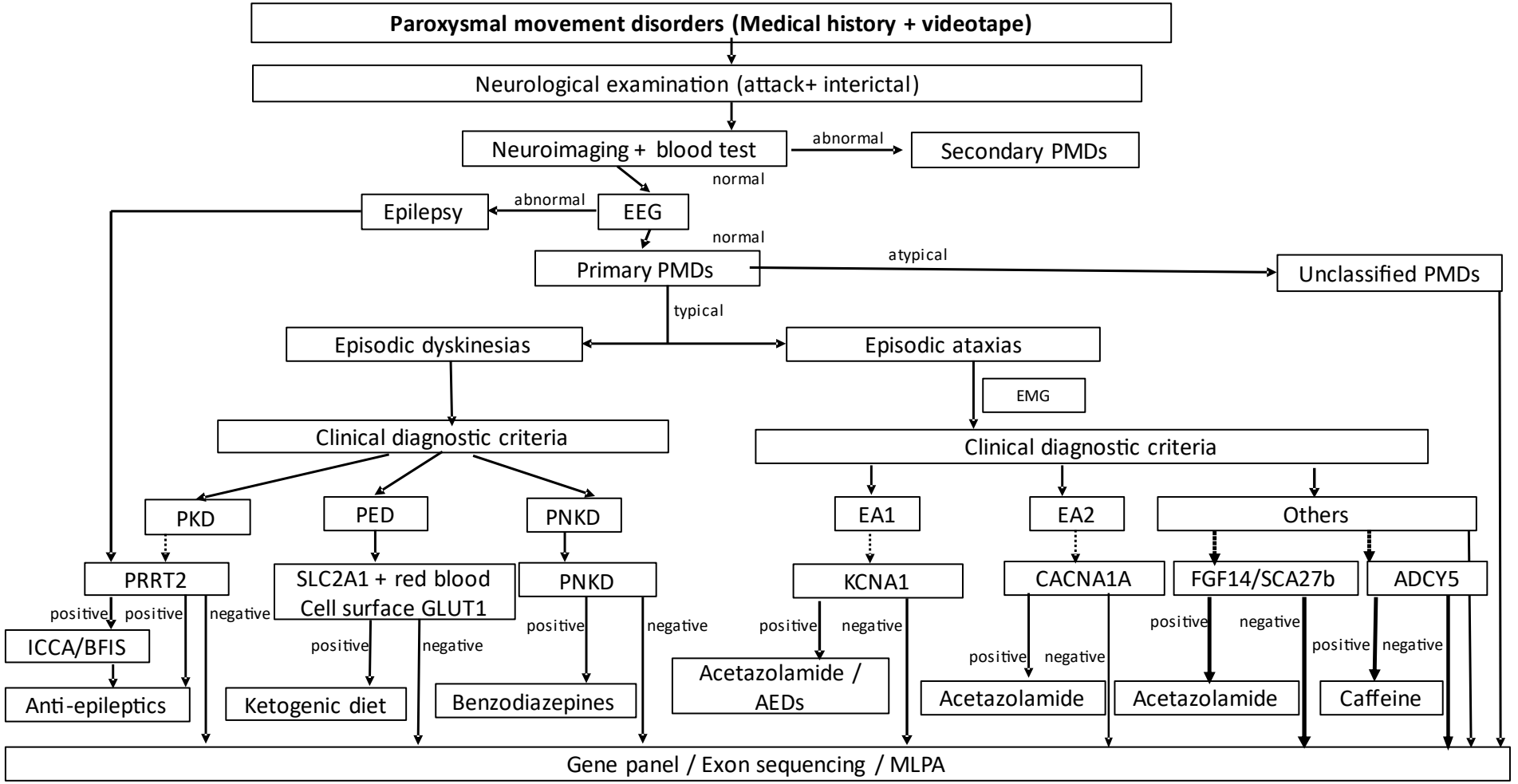
Flowchart development process:

- 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
- Revision of flowchart – October 2024 – October 2025
- Consent on revision by whole disease group – 27 October 2025

DIAGNOSTIC FLOWCHART FOR PAROXYSMAL DISORDERS

Adapted from [Zhang XJ, Xu ZY, Wu YC, Tan EK. Paroxysmal movement disorders: Recent advances and proposal of a classification system. Parkinsonism Relat Disord. 2019 Feb;59:131-139. doi: 10.1016/j.parkreldis.2019.02.021. Epub 2019 Feb 18. PMID: 30902529]



ABBREVIATIONS

ADCY5	–	Adenylate Cyclase 5
AEDs	–	Anti-Epileptic Drugs
CACNA1A	–	Calcium Voltage-Gated Channel Subunit Alpha-1A
EA1	–	Episodic Ataxia Type 1
EA2	–	Episodic Ataxia Type 2
EEG	–	Electroencephalography
EMG	–	Electromyography
FGF14	–	Fibroblast Growth Factor 14
GLUT1	–	Glucose Transporter Type 1
ICCA/BFIS	–	Infantile Convulsions with Choreoathetosis / Benign Familial Infantile Seizures
KCNA1	–	Potassium Voltage-Gated Channel Subfamily A Member 1
MLPA	–	Multiplex Ligation-Dependent Probe Amplification
SCA27b	–	Spinocerebellar Ataxia Type 27b
SLC2A1	–	Solute Carrier Family 2 Member 1
PED	–	Paroxysmal Exercise-Induced Dyskinesia
PKD	–	Paroxysmal Kinesigenic Dyskinesia
PMDs	–	Paroxysmal Movement Disorders
PNKD	–	Paroxysmal Non-Kinesigenic Dyskinesia
PRRT2	–	Proline-Rich Transmembrane Protein 2

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