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 ATYPICAL PARKINSONISM AND GENETIC PD

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW PREVALENCE AND COMPLEX DISEASES

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Published by ERN-RND: 9th January 2019
Updated on 20th March 2026

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Luxembourg: Publications Office of the European Union, 2019

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, the 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 Disorders and Neurodegeneration with Brain Iron Accumulation
- Frontotemporal Dementia
- Huntington's Disease and other Choreas
- Leukoencephalopathies

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

Recommendation for clinical use:
The European Reference Network for Rare Neurological Diseases developed the Diagnostic Flowchart for atypical parkinsonism and genetic Parkinson's disease to help guide the diagnosis. The ERN-RND recommends the use of this Diagnostic Flowchart.

DISCLAIMER

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METHODOLOGY

The Diagnostic Flowchart was developed and revised by the Disease Group for Atypical Parkinsonism and Genetic PD of ERN-RND

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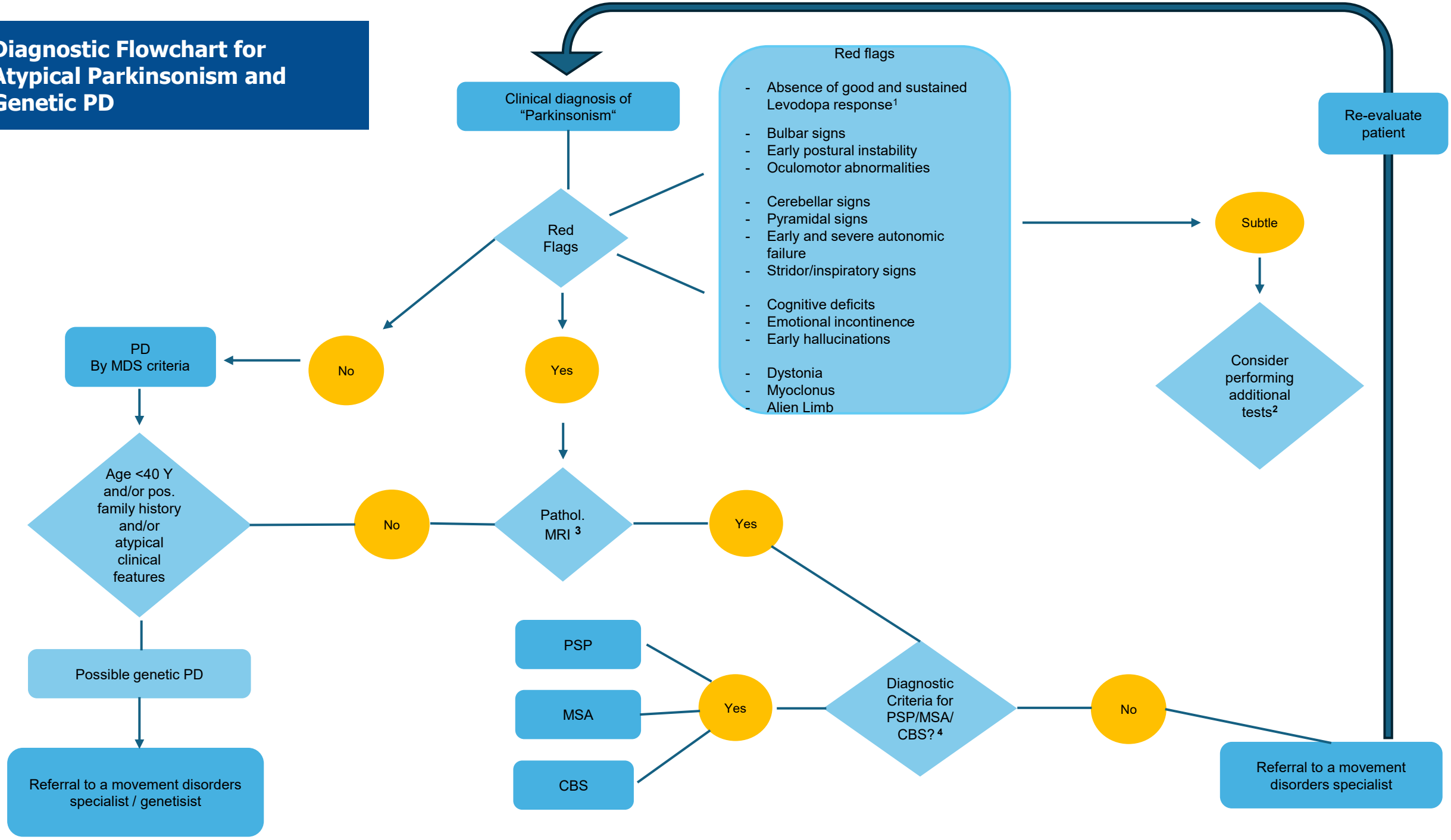
Flowchart development process:

- Development of flowchart – June – November 2017
- Discussion/Revision in ERN-RND disease – November 2017 – June 2018
- Consent on diagnostic flowchart during ERN-RND annual meeting 2018 – 08/06/2018
- Consent on document by whole disease group – 15/11/2018
- Proposal to Disease Group to revise Diagnostic Flowchart – 11/10/2024
- Revision of Diagnostic Flowchart – October 2024 - February 2026
- Final consent on revised Diagnostic Flowchart – 18/03/2026
- Publication of revised document on ERN-RND webpage – 20/03/2026

Diagnostic Flowchart for Atypical Parkinsonism and Genetic PD



1. Good and sustained Levodopa response:

- Levodopa Challenge (200 mg Levodopa) with a >30% improvement of the UPDRS III
- If the improvement is <30% UPDRS III , increase the Levodopa Dosis or try a chronic therapy over at least 3 months
- Good response for >1 year

2. Additional tests:

- Bladder ultrasound/Urodynamic test
- Schellong test/Tilt test
- Videooculography
- Neuropsychological Evaluation
- Fibroendoscopic evaluation of swallowing (FEES)
- Polysmonograhy

3. Pathological MRI signs:

- Atrophy of putamen, middle cerebellar peduncle, pons, cerebellum
- Hot cross bun sign
- Increase diffusivity of putamen/middle cerebellar peduncle
- Predominant midbrain atrophy
- Asymmetric cortical atrophy of perirolandic region, posterior frontal and parietal lobe

4 Diagnostic criteria:

CBD: Armstrong, M. J., et al. (2013). "Criteria for the diagnosis of corticobasal degeneration." Neurology 80(5): 496–503.

PSP: Hoglinger, G. U., et al. (2017). "Clinical diagnosis of progressive supranuclear palsy: The movement disorder society criteria." Mov Disord 32(6): 853–864.

MSA: Wenning, G. K., et al. (2022). "The Movement Disorder Society Criteria for the Diagnosis of Multiple System Atrophy." Mov Disord.

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Co-funded by the European Union