European Reference Networks

European Reference Network for rare or low prevalence complex diseases

Network: Neurological Diseases (ERN-RND)

Coordinator: Universitätsklinikum Tübingen — Deutschland

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 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 centres and the diseases covered can be found on the network website 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

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.

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METHODS

The development of the Diagnostic Flowcharts for Dystonia has been performed by the Disease group for Dystonia, paroxysmal disorders and NBIA of ERN-RND.

Consent on revised document was given by the entire Disease Group on: 27 October 2025

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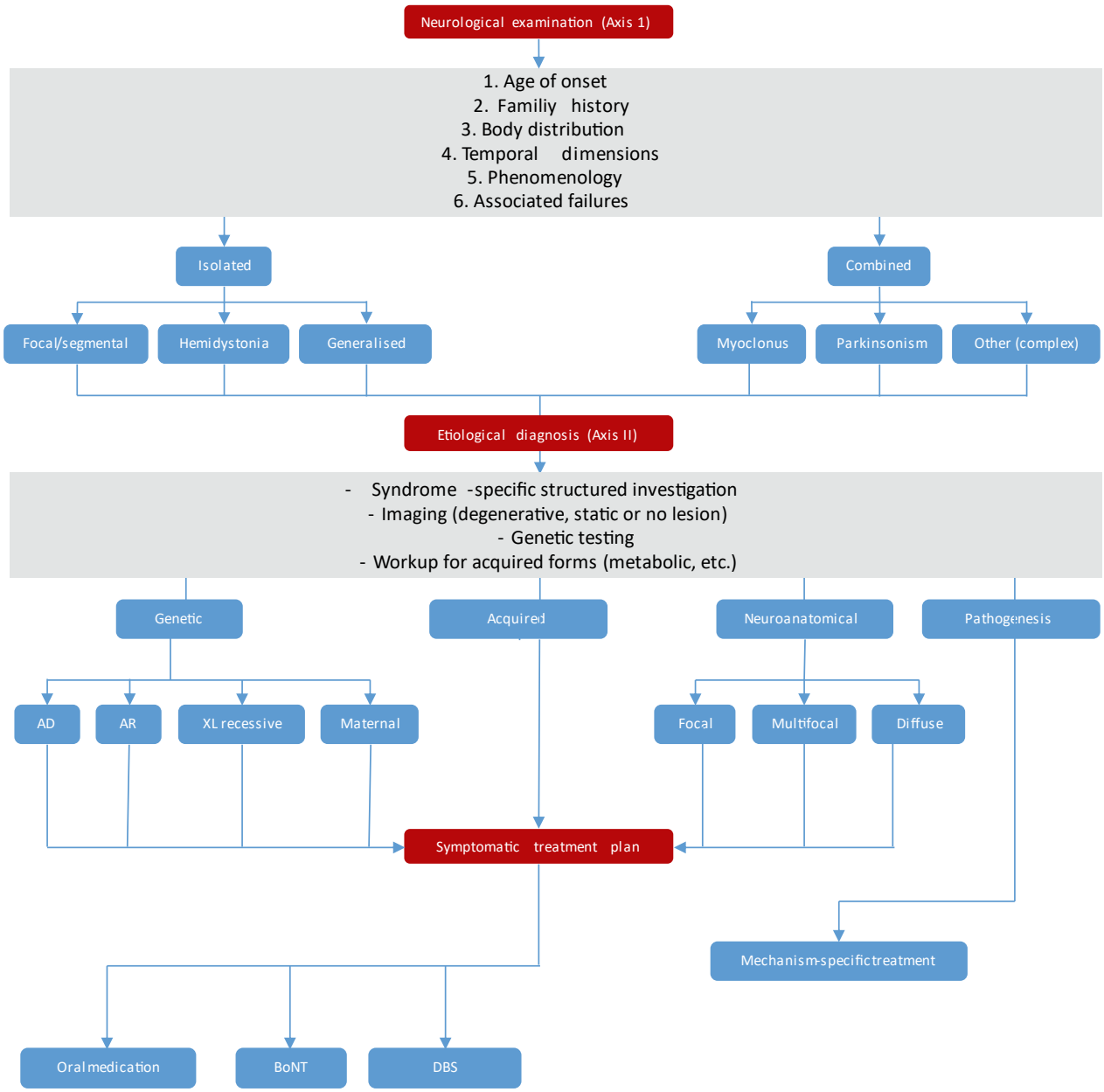
Endorsement process:

- 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 Diagnostic Flowchart for Dystonia in adults – October 2024 – October 2025
- Consent on revision by whole disease group – 27/10/2025-

Diagnostic flowchart for Dystonia in adults

Adapted and compiled in accordance to

- Albanese A, Bhatia K, Fung VSC, Hallett M, Jankovic J, Jinnah HA, Klein C, Lang AE, Mink JW, Pandey S, Teller JK, Tijssen MAJ, Vidailhet M, Jinnah HA. Phenomenology and classification of dystonia. Mov Disord. 2025 in press. doi: 10.1002/mds.30220. Epub 2025 May 6. PMID: 40326714.
- Albanese, A., Di Giovanni, M. and Lalli, S. (2019), Dystonia: diagnosis and management. Eur J Neurol, 26: 5-17. https://doi.org/10.1111/ene.13762
- Grütz K, Klein C. Dystonia updates: definition, nomenclature, clinical classification, and etiology. J Neural Transm (Vienna). 2021 Apr;128(4):395-404. doi: 10.1007/s00702-021-02314-2. Epub 2021 Feb 19.
- Stephen CD. The Dystonias. Continuum (Minneap Minn). 2022 Oct 1;28(5):1435-1475. doi: 10.1212/CON.0000000000001159. PMID: 36222773; PMCID: PMC10226676.



Abbreviations:

AD: Autosomal dominant
AR: Autosomal recessive
BoNT: Botulinum Neurotoxin Treatment
DBS: Deep Brain Stimulation
XL: X-(Chromosome-)linked

Principal Monogenic Dystonia (Axis II)

Gene	Old Name	Transmission	Neurological Examinaiton (Axis I)
DYT-ANO3	DYT-24	AD	**Isolated dystonia** affecting neck, laryngeal muscles and upper limbs with tremor.
DYT-EIF2AK2 (LEUDEN)		AD	**Isolated dystonia** with generalised involement, often accompanied by leukoencephalopathy, developmental delay, and episodic neurologic regression syndrome (LEUDEN).
DYT-GNAL	DYT-25	AD	**Isolated dystonia** with adult onset of focal dystonia usually involing the neck.
DYT-KMT2B	DYT-28	AD	**Isolated dystonia** with focal involvement of lower limb and secondary progression. First decade. Occasionally developemental delay.
DYT-THAP1	DYT-6	AD	**Isolated dystonia** with involvement of craniofacial muscles with secondary diffusion. Adolescent-onset dystonia.
DYT-TOR1A	DYT-1	AD	**Isolated dystonia** with torsion of the neck, the trunk or limbs. Early-onset (first decade).
DYT-VPS16	DYT-30	AD	**Isolated dystonia** oromandibular, cervical, bulbar or upper limb dystonia.
DYT-PRKRA	DYT-16	AR	**Isolated dystonia** early onset dystonia with **parkinsonism**, with dysphagia and spasmodic dystonia, torticolli and upper limb dystonia.
DYT-DNAJC12		AR	**Combined dystonia** with hyperphenylalaninemia, developemental delay, **parkinsonism** levodopa-responsive.
DYT/PARK-TAF1	DYT-3	XL	**Combined dystonia – parkinsonsim** involving the eye blinking, neck, upper or lower limbs.
DYT/PARK-TH		AR	**Combined dystonia – parkinsonsim** with generalised involvement levodopa-responsive. 3 different phenotypes with or without encephalopathy.
DYT/PARK-ATP1A3	DYT-12	AD	**Combined dystonia – parkinsonsim** asymmetic and with rapid onset. Often bulbar disfunction. Chorea in later life.
MYC/DYT-KCTD17	DYT-26	AD	**Combined dystonia – myoclonus** with predominant involvement of cranal and laryngeal muscle.
MYC/DYT-SGCE	DYT-11	AD	**Combined dystonia – myoclonus** with torticollis and wrtier‘s cramp with tremor.

MDS Complete List of Monogenic Dystonias

Isolated dystonias

DYT-ANO3
DYT-EIF2AK2
DYT-GNAL
DYT-HPCA
DYT-KMT2B
DYT-PRKRA
DYT-THAP1
DYT-TOR1A
DYT-VPS16

Combined dystonias

DYT-COX20
DYT-DNAJC12
DYT-SLC39A14
DYT/PARK-ATP1A3
DYT/PARK-GCH1
DYT/PARK-TAF1
DYT/PARK-TH
DYT/CHOR-GNAO1
MYC/DYT-KCTD17
MYC/DYT-SGCE

Complex dystonia (where dystonia dominates the clinical picture but this occurs in the context of a complex phenotype including symptoms other than movement disorders)

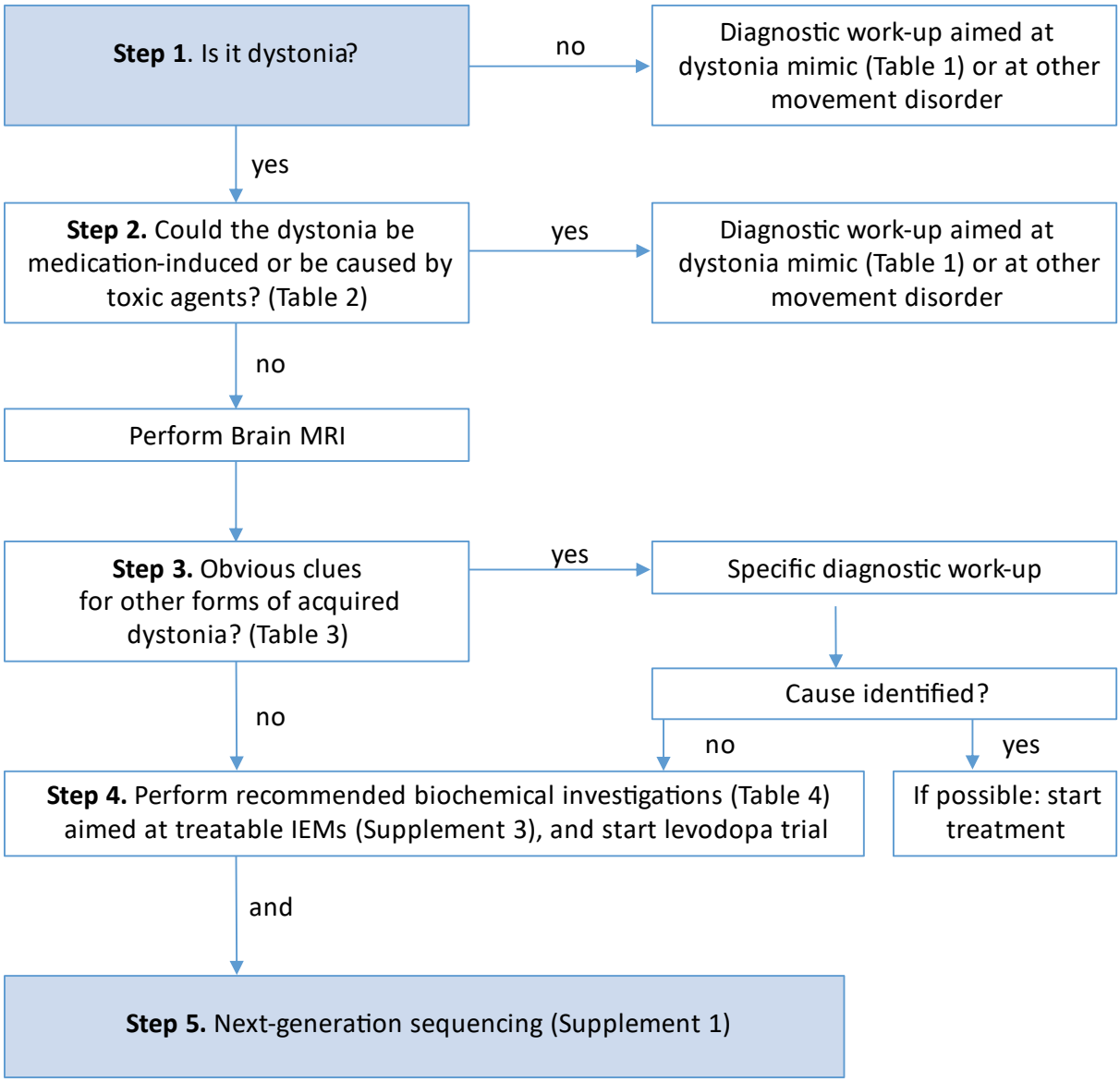
DYT-ACTB
DYT-ATP7B
DYT-BCAB31
DYT-DCAF17-(NBIA)
DYT-DDC
DYT-FITM2
DYT-IRF2BPL
DYT-MECR
DYT-mt-ND6
DYT-OPA1
DYT-PANK2 -(NBIA)
DYT-SERAC1
DYT-SLC19A3
DYT-SUCLA2
DYT-TIMM8A
DYT-TUBB4A
DYT-VAC14
DYT/CHOR-ACAT1
DYT/CHOR-ADAR1
DYT/CHOR-FOXG1
DYT/CHOR-GCDH
DYT/CHOR-HPRTDYT/CHOR-MUT
DYT/CHOR
PCCA/PCCB
DYT/PARK-CP-(NBIA)
DYT/PARK-GLB1
DYT/PARK-PLA2G6-(NBIA)
DYT/PARK-PTS
DYT/PARK-QDPR
DYT/PARK-SCL6A3
DYT/PARK-SCL30A10
DYT/PARK-SPR
ATX/DYT-SQSTM1

Disorders that usually present with other phenotypes but can have predominant dystonia

ATX-ATXN3
HSP-C19orf12-(NBIA)
HSP/ATX-FA2H-(NBIA)
HSP/ATX-KIF1C
CHOR-FTL-(NBIA)

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



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