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 EARLY-ONSET ATAXIAS

Consented by ERN-RND: July 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 (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 32 of Europe's leading expert centres as well as 10 affiliated partners in 20 member states and includes highly active patient organizations. Centres are located in Austria, Belgium, Bulgaria, Czech Republic, Denmark, Estonia, Finland, France, Germany, Hungary, Italy, Latvia, Lithuania, Luxembourg, Malta, 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 Parkinson's disease
- Dystonia, Paroxysmal Disorder and Neurodegeneration with Brain Ion 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 networks web site www.ern-rnd.eu.

Recommendation for clinical use:
The European Reference Network for Rare Neurological Diseases developed the Diagnostic Flowchart for early-onset Ataxias to help guide the diagnosis. The Reference Network recommends the use of this Diagnostic Flowchart.

DISCLAIMER

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METHODOLOGY

The development of the Diagnostic Flowchart was done by the Disease group for Ataxia and Hereditary Spastic Paraplegias of ERN-RND.

Disease group for Ataxia and Hereditary Spastic Paraplegias:

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Flowchart development process:

- Development of flowchart – September 2019 – July 2020
- Consent on document by whole disease group – July 2020

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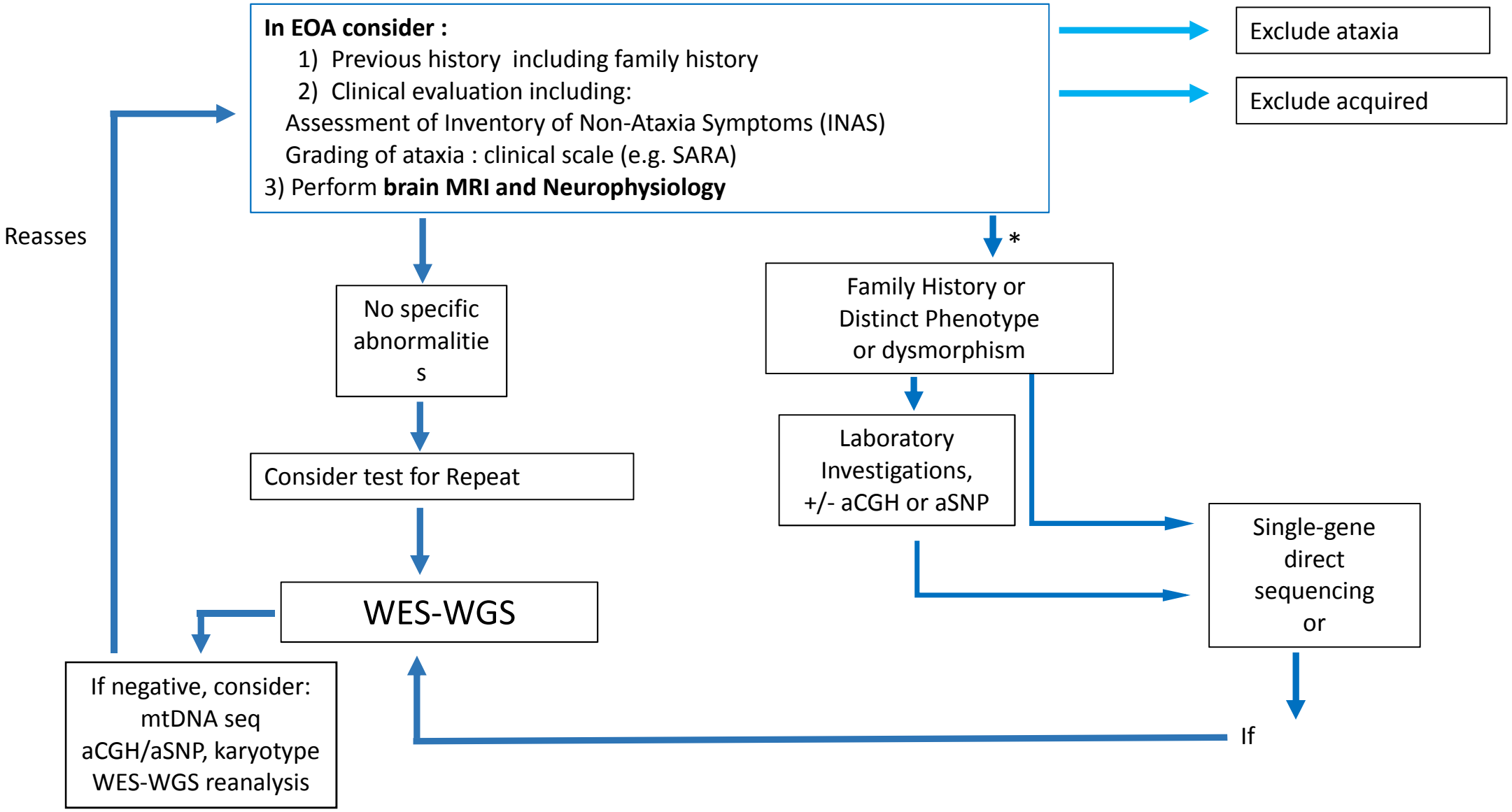
Early Onset Ataxia



Notes:
WES-WGS could be initiated, if timely available, parallel to the investigations following brain MRI
*Distinct EOA phenotypes and targeted investigations reviewed in Brandsma R et al., Eur J Paediatr Neurol 2019

Abbreviations:
aCGH – microarray-based Comparative Genomic Hybridisation
aSNP – microarray-based testing for Single Nucleotide Polymorphisms
mtDNA seq – Mitochondrial DNA sequencing
MRI – Magnetic Resonance Imaging
NGS – Next Generation Sequencing
WES – Whole Exome Sequencing
WGS – Whole Genome Sequencing

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

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