



# Diagnostic flowchart for adult ataxias

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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 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 Parkinson's disease
- Dystonia, Paroxysmal Disorder and Neurodegeneration with Brain Ion Accumulation
- Frontotemporal Dementia
- Huntingtons' Disease and other Chorea
- Leukodystrophies

Specific information about the network, the expert centres and the diseases covered can be found at the networks web site [www.ern-rnd.eu](http://www.ern-rnd.eu).

## Recommendation for clinical use:

**The European Reference Network for Rare Neurological Diseases developed the Diagnostic Flowchart for adult 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:

### **Disease group coordinators:**

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Patient representatives:

Lori Renna Linton<sup>10</sup>, Mary Kearney<sup>10</sup>, Cathalijne van Doorne<sup>10</sup>

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## Flowchart development process:

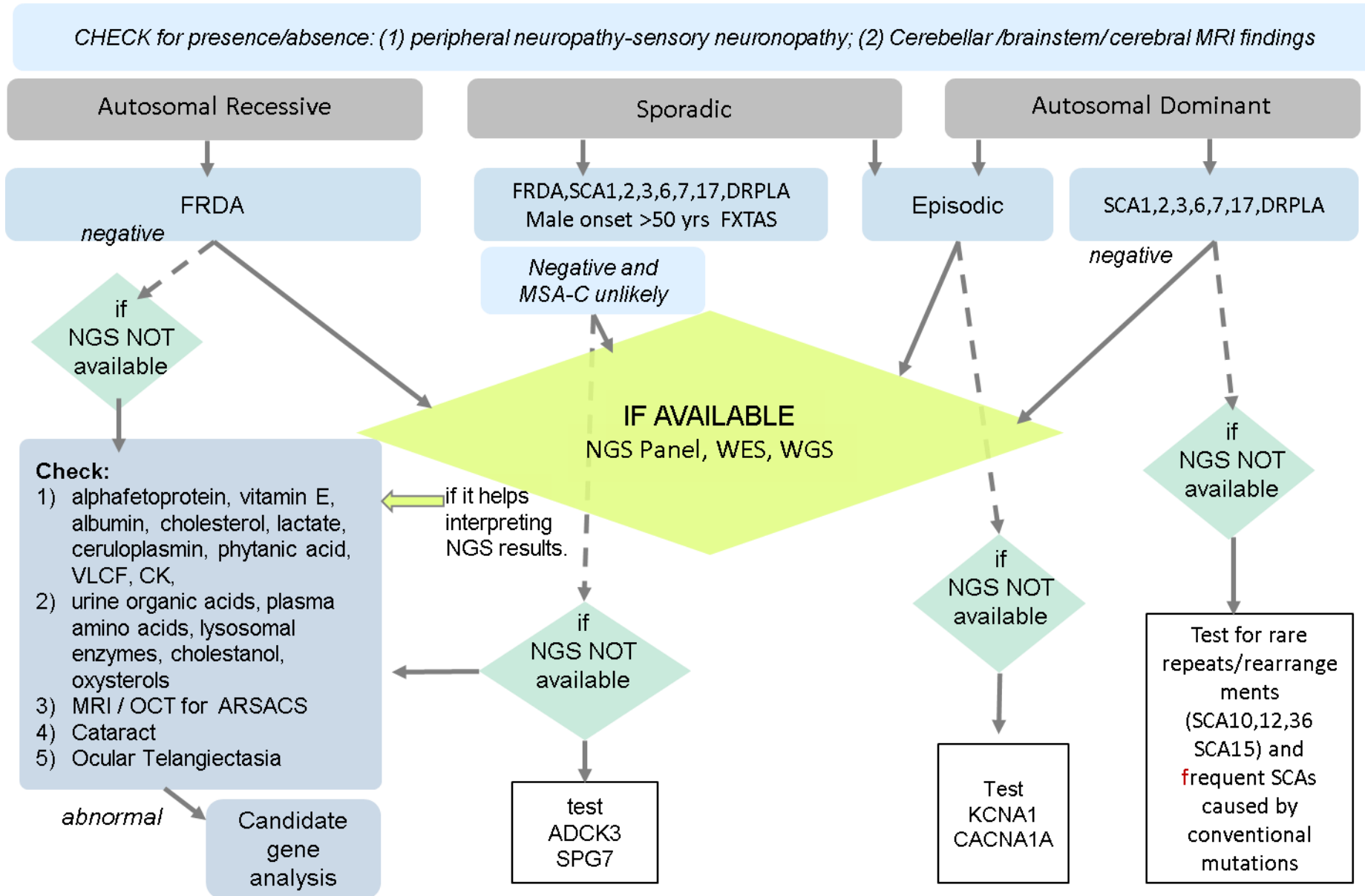
- Development of flowchart – June – November 2017
- Discussion/Revision in ERN-RND disease group – November 2017 – June 2018
- Consent on diagnostic flowchart:30 November 2018
- Consent on document by whole disease group – 05/02/2019



# Diagnostic flowcharts –Ataxias

Exclusion of acquired causes\*\*\* in case of negative family history, (sub)acute onset, specific medical history, etc.

\*\*\*Common Acquired Causes: auto immune diseases (MS, sarcoidosis, celiac disease, etc), toxic reaction, head trauma, cerebral palsy, tumor, stroke, infections, vitamine deficiency, paraneoplastic syndromes



## **ABBREVIATIONS**

ADCK3: aarF domain-containing protein kinase 3

ARSACS: autosomal recessive spastic ataxia of Charlevoix-Saguenay

CACNA1A: Calcium Voltage-Gated Channel Subunit Alpha1 A

CK: creatine kinase

DRPLA: Dentatorubral-pallidoluysian atrophy

FRDA: friedreich ataxia

FXTAS: fragile X-associated tremor/ataxia syndrome

KCNA1: Potassium Voltage-Gated Channel Subfamily A Member 1

MRI: Magnetic Resonance Imaging

MSA-C: multiple system atrophy, cerebellar type

NGS: next-generation sequencing

OCT: optical coherence tomography

SCA: spinocerebellar ataxia

SPG7: spastic paraplegia type 7

VLCF: very long-chain fatty acids

WES: whole-exome sequencing

WGS: whole-genome sequencing





# European Reference Networks

[https://ec.europa.eu/health/ern\\_en](https://ec.europa.eu/health/ern_en)



## European Reference Network

for rare or low prevalence  
complex diseases

⚙️ **Network**  
Neurological Diseases  
(ERN-RND)

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