

Diagnosis and management of genetic ataxias in adulthood: an EAN/ERN-RND guideline

DG/WG
Ataxia & HSPs



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Background & Rationale

The pooled prevalence of genetic ataxias in Europe was estimated to be as high as 11.2 cases per 10,000 persons in 2014 (absolute prevalence: 831,376 cases). The true estimate is likely to be much higher. In fact, recent advances in molecular genetics have shown that several sporadic ataxias do indeed have a genetic cause.

The choice of optimal diagnostic procedures in genetic ataxias is complicated by the pleiotropy of > 100 ataxia genes, the wide phenotypic variability and the multiple underlying mutational mechanisms, which require prioritization of genetic testing strategies.

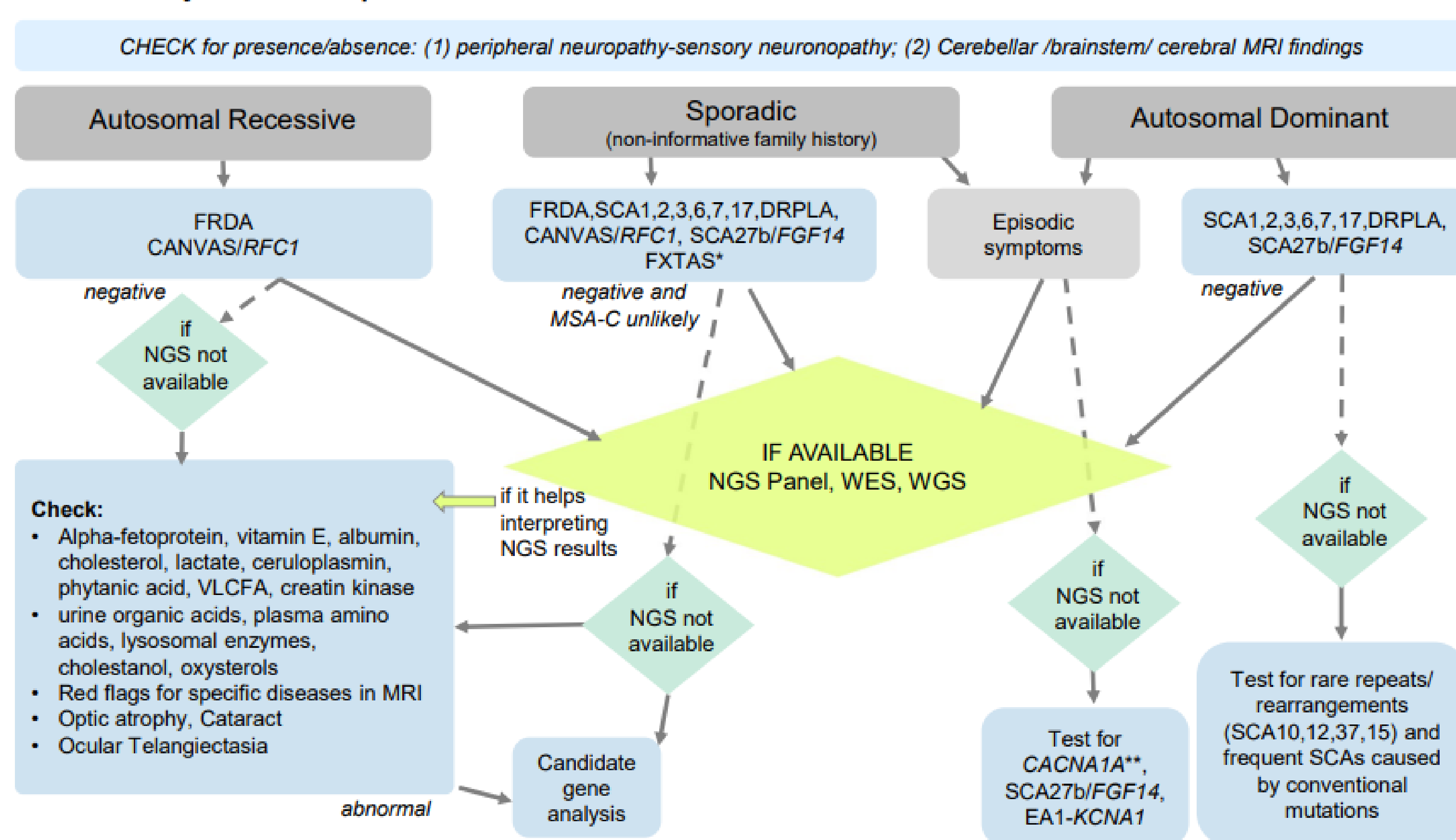
Genetic diagnostic practices vary widely across Europe due to financial and availability issues. Nevertheless, first-line diagnostics in genetic ataxias can be performed with widely available, low-cost diagnostic tools before applying next-generation sequencing or long-read sequencing.

In terms of clinical management, there has been some exciting news in recent years. The first treatment for Friedreich's ataxia, was approved in the European Union in 2024, making early diagnosis imperative. Evidence of efficacy has also accumulated in recent years for several other therapeutic approaches (e.g. 4-aminopyridine in GAA-FGF14 ataxia, N-acetyl-l-leucine in Niemann-Pick type C disease).

Improving the diagnostic pathway in adult Ataxias 1: An updated Flowchart

Exclusion of acquired causes in case of (sub)acute onset, specific medical history or MRI findings. Common acquired causes include: autoimmune diseases, toxins, head trauma, hypoxia, tumor, stroke, infections, vitamin deficiency, paraneoplastic syndromes

N.B.: For early-onset ataxias please consult the dedicated ERN-RND flowchart.



See also the Educational Webinar by Enrico Bertini & Elisabetta Indelicato
<https://www.youtube.com/watch?v=B0QEIQtjMFU>

Improving the diagnostic pathway in adult Ataxias 2: An updated Guideline

An updated consensus flow chart for the diagnosis of adult ataxias has recently been published by the ERN-RND. However, the lack of an accompanying publication limits its use and visibility.

The last European consensus paper on the diagnosis and treatment of adult genetic ataxia from the EFNS/ENS was published 11 years ago. Recent advances in the diagnosis and treatment of genetic ataxia make an updated guideline on this topic urgently needed. Advances in term of treatments in the previous five years make likewise an early diagnosis a priority.

Objectives & Outlook

- Updated guidelines for the diagnosis and management of adult genetic ataxia are urgently needed.
- This project fits with the ERN-RND goal of improving standards of care, accelerating diagnosis and potentially improving access to therapy for patients with ataxia.
- Given the general lack of such a comprehensive publication in the literature, such guidelines have the potential to become the reference text on the subject at a global level.

Submission to the EAN Guideline Committee and selection as „priority topic“ for the period 2025-2027

Request for funding submitted and approved by the ERN-RND

Next step:
Definition of key guideline data within the present ERN-RND meeting and in a first online kick-off meeting

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