

DIAGNOSTIC FLOWCHART FOR ADULT ATAXIAS

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW PREVALENCE AND COMPLEX DISEASES

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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, 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 Disorder and Neurodegeneration with Brain Iron Accumulation
- Frontotemporal Dementia
- Huntington's Disease and other Chorea
- Leukodystrophies

Specific information about the network, the expert centers and the covered diseases can be found on the network's website 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.

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METHODOLOGY

The development of the Diagnostic Flowcharts for Adult Ataxias 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:

Elisabetta Indelicato²⁷; Lorenzo Nanetti¹⁹; Rebecca Schuele-Freyer³⁸

Disease Group members:

Aki Hietaharju¹⁴; Alejandra Darling¹⁷; Alena Zumrová²⁸; Alexander Münchau⁴¹; Alexandra Durr⁶; Alfons Macaya⁴⁸; Ana Lara Pelayo²⁶; Andrea Mignarri⁵; Andrés Nascimento¹⁷; Angelo Antonini³; Anna Ardissoni¹⁹; Anna Fetta²⁰; Anna Heinzmann⁶; Anna Sobanska¹⁸; Anna Sulek¹⁸; Antonio Federico⁵; Aoife Mahony²¹; Astrid Daniela Adarmes Gómez⁴⁹; Bart van de Warrenburg³¹; Bela Melegh⁴⁷; Berry Kremer⁴³; Borut Peterlin⁴⁵; Carlos Orteza¹⁷; Caroline Scicluna⁶; Caterina Garone²⁰; Caterina Mariotti¹⁹; Charlotte Haaxma³¹; Chiara Criscuolo²; Chiara Pane²; Christa-Caroline Bergner⁴⁶; Claire Ewencyk⁶; Colin Clarke²¹; Damjan Osredkar⁴⁵; Daniel Boesch²⁷; David Gómez-Andrés⁴⁸; David Neubauer⁴⁵; Deborah Sival⁴³; Duccio Maria Cordelli²⁰; Eleni Zamba-Papanicolaou¹⁰; Enrico Bertini³⁰; Esteban Muñoz¹⁷; Eugenia Amato¹⁷; Fran Borovecki³⁷; Francesco Nicita³⁰; Francisco Javier Rodríguez de Rivera²³; Franco Taroni¹⁹; Gabriele Siciliano⁴; Georgia Karadima¹¹; Georgios Koutsis¹¹; Gessica Vasco³⁰; Gilles Naeije¹³; Ginevra Zanni³⁰; Giovanna De Michele²; Giovanni Rizzo²⁰; Giulia Coarelli⁶; Harri Rusanen¹⁴; Helena Sarac³⁷; Hendrik Rosewich⁴²; Ieva Glazere²⁹; Isabella Moroni¹⁹; Iwona Stępnia¹⁸; Jan Lykke Scheel Thomsen¹; Jeroen Vermeulen²⁵; Jiri Klempir¹⁵; Joanna Pera³⁹; Jon Infante²⁶; Judith van Gaalen³¹; Kathleen Gorman²¹; Kevin Peikert⁴⁴; Kinga Hadziev⁴⁷; Krista Lazdovska²⁹; Kristl Claeys⁴⁰; Kyproula Christodolou¹⁰; Laszlo Szpisjak³⁴; Laura Carrera¹⁷; Laurine Virchien⁹; Lena Hjerminde³²; Luca Solina²⁰; Lucie Pierron⁶; Ludger Schöls⁴²; Mar O'Callaghan¹⁷; Marcus Grobe-Einsler³⁶; Margit Lill³⁵; Maria Victoria Gonzalez Martinez⁴⁸; Marit Otto¹; Marta Blázquez Estrada⁷; Marta Correa⁴⁹; Martin Paucar Arce²²; Martin Vyhnálek²⁸; Martje Pauly⁴¹; Matthis Synofzik⁴²; Maurizio Petrarca³⁰; Michael Bayat¹; Michèl Willemsen³¹; Michelangelo Mancuso⁴; Miryam Carecchio³; Norbert Brüggemann⁴¹; Norbert Kovacs⁴⁷; Pablo Mir⁴⁹; Perrine Charles⁶; Peter Balicza³³; Péter Klivényi³⁴; Peter Martus⁴²; Petya Bogdanova-Mihaylova²¹; Pierre Kolber⁸; Rachele Danti¹⁹; Ramona Valante²⁹; Richard Walsh²¹; Sára Davisonová¹⁵; Sinead Murphy²¹; Susanne Petri¹⁶; Susanne Schneider²⁴; Sylvia Boesch²⁷; Veronica Di Pisa²⁰; Viktor Molnar³³; Vincenzo Montano⁴; Virginie Destrebecq¹³; Wolfgang Koehler⁴⁶; Wolfgang Nachbauer²⁷; Yorck Hellenbroich⁴¹

Patient representative:

John Gerbild¹²; Juliane Krabath¹²; Lori Renna Linton¹²; Mary Kearney¹²

¹Aarhus University Hospital, Denmark; ²AOU - Federico II University Hospital, Naples, Italy; ³AOU - University Hospital Padua, Italy; ⁴AOU - University Hospital Pisa, Italy; ⁵AOU - University Hospital Siena, Italy; ⁶APHP - Reference Centre for Rare Diseases 'Neurogenetics', Pitié-Salpêtrière Hospital, Paris, France; ⁷Asturias Central University Hospital, Oviedo, Spain; ⁸CHL - Luxembourg Hospital Center, Luxembourg; ⁹CHU de Toulouse, France; ¹⁰Cyprus Institute of Neurology and Genetics, Egkomi, Cyprus; ¹¹Eginitio Hospital, National and Kapodistrian University of Athens, Greece; ¹²ePAG representative; ¹³Erasmus Hospital, Université libre de Bruxelles, Brussels, Belgium; ¹⁴Finland Consortium: University Hospitals in Oulu, Tampere and Helsinki, Finland; ¹⁵General University Hospital Prague, Czech Republic; ¹⁶Hannover Medical School, Germany; ¹⁷Hospital Clinic Barcelona and Sant Joan de Déu Hospital,

Barcelona, Spain; ¹⁸Institute of Psychiatry and Neurology, Warsaw, Poland; ¹⁹IRCCS - Foundation of the Carlo Besta Neurological Institute, Milan, Italy; ²⁰IRCCS - Institute of Neurological Sciences of Bologna, Italy; ²¹Irish Consortium: Tallaght University Hospital and Children's Health Ireland, Dublin, Ireland; ²²Karolinska University Hospital, Stockholm, Sweden; ²³La Paz University Hospital, Madrid, Spain; ²⁴Ludwig-Maximilian University Hospital, Munich, Germany; ²⁵Maastricht University Medical Center, Netherlands; ²⁶Marques de Valdecilla University Hospital, Santander, Spain; ²⁷Medical University Innsbruck, Austria; ²⁸Motol University Hospital, Prague, Czech Republic; ²⁹Pauls Stradins Clinical University Hospital, Riga, Latvia; ³⁰Pediatric Hospital Bambino Gesù, Rome, Italy; ³¹Radboud University Medical Centre, Nijmegen, Netherlands; ³²Rigshospitalet University Hospital Copenhagen, Denmark; ³³Semmelweis University, Budapest, Hungary; ³⁴Szent-Györgyi Albert Medical Center, Szeged, Hungary; ³⁵Tartu University Hospital, Estonia; ³⁶University Hospital Bonn, Germany; ³⁷University Hospital Center Zagreb, Croatia; ³⁸University Hospital Heidelberg, Germany; ³⁹University Hospital in Krakow, Poland; ⁴⁰University Hospital Leuven, Belgium; ⁴¹University Hospital Schleswig-Holstein, Germany; ⁴²University Hospital Tübingen, Germany; ⁴³University Medical Center Groningen, Netherlands; ⁴⁴University Medical Center Rostock, Germany; ⁴⁵University Medical Centre Ljubljana, Slovenia; ⁴⁶University of Leipzig Medical Center, Germany; ⁴⁷University of Pécs, Hungary; ⁴⁸Vall d'Hebron University Hospital, Barcelona, Spain; ⁴⁹Virgen del Rocío University Hospital, Sevilla, Spain

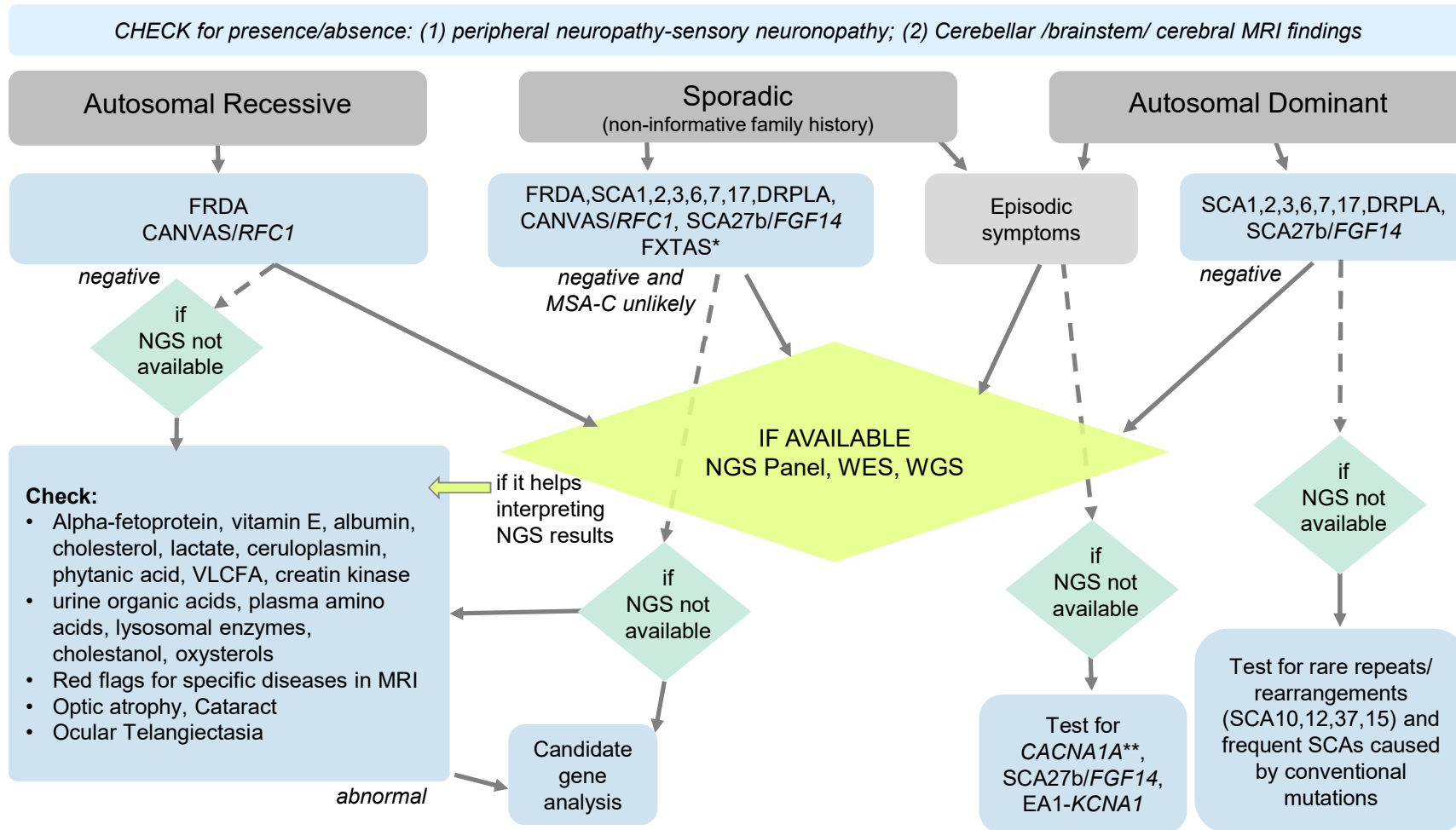
Flowchart development process:

- Development of flowchart: June – November 2017
- Discussion 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
- Revision of flowchart: February 2024
- Consent on Revision by whole disease group: September 2024

Diagnostic flowchart –Ataxias

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, vitamine deficiency, paraneoplastic syndromes

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



Notes:

* Consider FXTAS if tremor-ataxia syndrome and age of onset >50 years

** Test for both conventional mutations (episodic ataxia type 2 and overlap syndromes hemiplegic migraine/episodic ataxia) and CAG expansions (SCA6 with early episodic symptoms)

Abbreviations:

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|--------|--|
| CANVAS | - Cerebellar Ataxia, Neuropathy, Vestibular Areflexia Syndrome |
| DRPLA | - Dentatorubral-pallidoluysian Atrophy |
| EA1 | - Episodic Ataxia type 1 |
| FRDA | - Friedreich's Ataxia |
| FXTAS | - Fragile X-associated Tremor/Ataxia Syndrome |
| MRI | - Magnetic Resonance Imaging |
| MSA-C | - Multiple System Atrophy Cerebellar type |
| NGS | - Next Generation Sequencing |
| SCA | - Spinocerebellar Ataxia |
| VLCFA | - Very Long Chain Fatty Acids |
| WES | - Whole Exome Sequencing |
| WGS | - Whole Genome Sequencing |



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 **Coordinator**
Universitätsklinikum
Tübingen — Deutschland

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