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 ADULT ATAXIAS

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW PREVALENCE AND COMPLEX DISEASES

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Published by ERN-RND: 11th February 2019

Revision published: 16th September 2024

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Luxembourg: Publications Office of the European Union, 2019

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, 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 Ion Accumulation
- Frontotemporal Dementia
- Huntingtons' Disease and other Choreas
- 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.

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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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 Indelicato[27]; Lorenzo Nanetti[19]; Rebecca Schuele-Freyer[38]

Disease Group members:

Aki Hietaharju[14]; Alejandra Darling[17]; Alena Zumrová[28]; Alexander Münchau[41]; Alexandra Durr[6]; Alfons Macaya[48]; Ana Lara Pelayo[26]; Andrea Mignarri[5]; Andrés Nascimento[17]; Angelo Antonini[3]; Anna Ardissone[19]; Anna Fetta[20]; Anna Heinzmann6; Anna Sobanska[18]; Anna Sulek[18]; Antonio Federico[5]; Aoife Mahony[21]; Astrid Daniela Adarmes Gómez[49]; Bart van de Warrenburg[31]; Bela Melegh[47]; Berry Kremer[43]; Borut Peterlin[45]; Carlos Ortez[17]; Caroline Scicluna[6]; Caterina Garone[20]; Caterina Mariotti[19]; Charlotte Haaxma[31]; Chiara Criscuolo[2]; Chiara Pane[2]; Christa-Caroline Bergner[46]; Claire Ewenczyk[6]; Colin Clarke[21]; Damjan Osredkar[45]; Daniel Boesch[27]; David Gómez-Andrés[48]; David Neubauer[45]; Deborah Sival[43]; Duccio Maria Cordelli[20]; Eleni Zamba-Papanicolaou[10]; Enrico Bertini[30]; Esteban Muñoz[17]; Eugenia Amato[17]; Fran Borovecki[37]; Francesco Nicita[30]; Francisco Javier Rodríguez de Rivera[23]; Franco Taroni[19]; Gabriele Siciliano[4]; Georgia Karadima[11]; Georgios Koutsis[11]; Gessica Vasco[30]; Gilles Naeije[13]; Ginevra Zanni[30]; Giovanna De Michele[2]; Giovanni Rizzo[20]; Giulia Coarelli[6]; Harri Rusanen[14]; Helena Sarac[37]; Hendrik Rosewich[42]; Ieva Glazere[29]; Isabella Moroni[19]; Iwona Stępniak[18]; Jan Lykke Scheel Thomsen[1]; Jeroen Vermeulen[25]; Jiri Klempir[15]; Joanna Pera[39]; Jon Infante[26]; Judith van Gaalen[31]; Kathleen Gorman[21]; Kevin Peikert[44]; Kinga Hadziev4[7]; Krista Lazdovska[29]; Kristl Claeys[40]; Kyproula Christodolou[10]; Laszlo Szpisjak[34]; Laura Carrera[17]; Laurine Virchien[9]; Lena Hjermind[32]; Luca Solina20; Lucie Pierron[6]; Ludger Schöls[42]; Mar O'Callaghan[17]; Marcus Grobe-Einsler[36]; Margit Lill[35]; Maria Victoria Gonzalez Martinez[48]; Marit Otto[1]; Marta Blázquez Estrada[7]; Marta Correa[49]; Martin Paucar Arce[22]; Martin Vyhnálek[28]; Martje Pauly[41]; Matthis Synofzik[42]; Maurizio Petrarca[30]; Michael Bayat[1]; Michèl Willemsen[31]; Michelangelo Mancuso[4]; Miryam Carecchio[3]; Norbert Brüggemann[41]; Norbert Kovacs[47]; Pablo Mir[49]; Perrine Charles[6]; Peter Balicza[33]; Péter Klivényi[34]; Peter Martus[42]; Petya Bogdanova-Mihaylova[21]; Pierre Kolber[8]; Rachele Danti[19]; Ramona Valante[29]; Richard Walsh[21]; Sára Davisonová[15]; Sinead Murphy[21]; Susanne Petri[16]; Susanne Schneider[24]; Sylvia Boesch[27]; Veronica Di Pisa[20]; Viktor Molnar[33]; Vincenzo Montano[4]; Virginie Destrebecq[13]; Wolfgang Koehler[46]; Wolfgang Nachbauer[27]; Yorck Hellenbroich[41]

Patient representative:

John Gerbild[12]; Juliane Krabath[12]; Lori Renna Linton[12]; Mary Kearney[12]

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

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

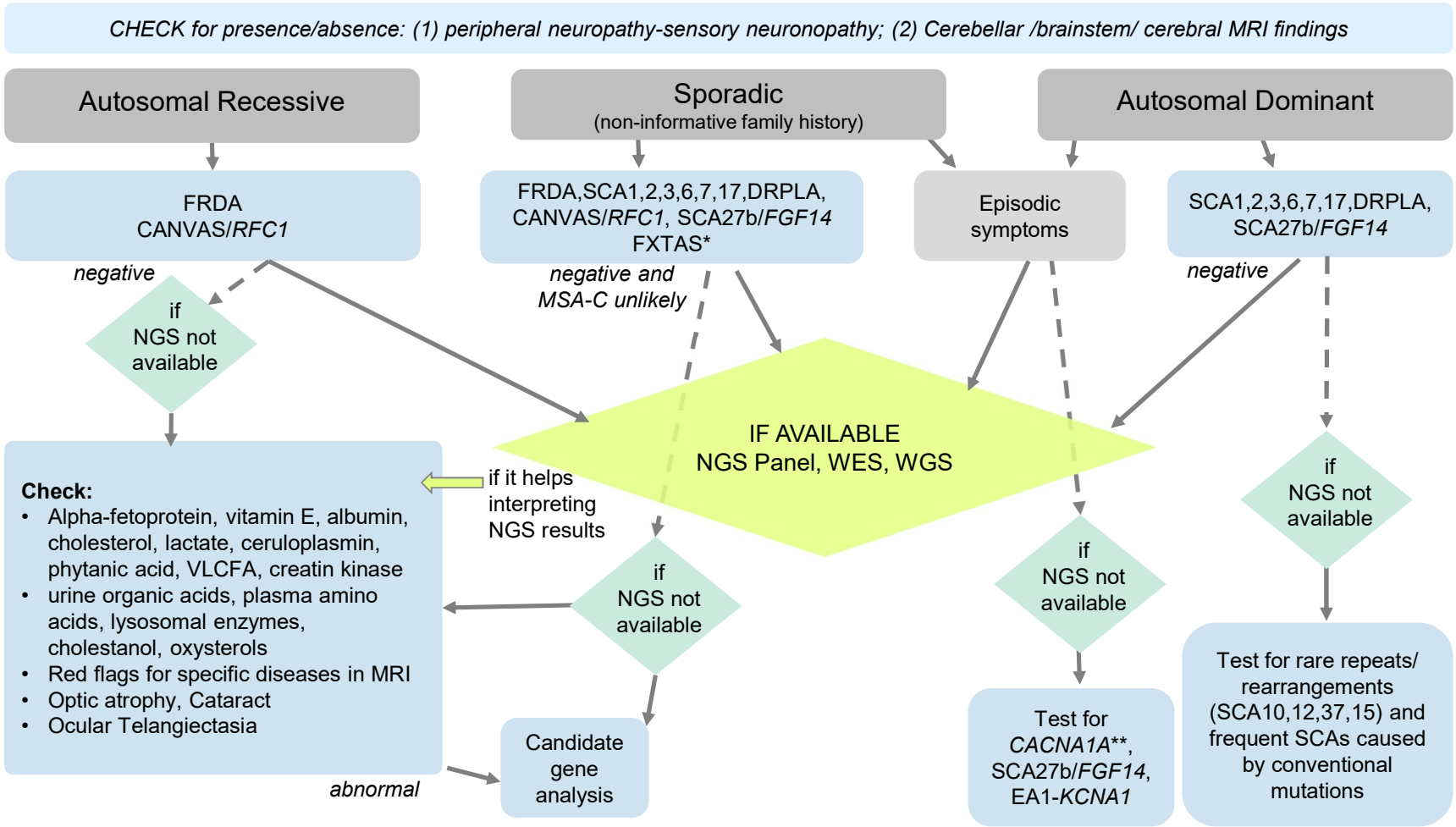
Flowchart development process:

- 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)

Abbrevations:

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

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

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Co-funded by the European Union