

Carola Reinhard<sup>1</sup>, Rebecca Schuele<sup>1</sup>, Caterina Mariotti<sup>2</sup>, Enrico Bertini<sup>3</sup>, Alfons Macaya<sup>4</sup>, Odile Boespflug-Tanguy<sup>5</sup>, Samuel Groeschel<sup>1</sup>, Ingeborg Krägeloh-Mann<sup>1</sup>, Nicole Wolf<sup>6</sup>, Rik Vandenberghe<sup>7</sup>, Isabelle Le Ber<sup>8</sup>, Markus Otto<sup>9</sup>, Tobias Baeumer<sup>10</sup>, Belén Pérez Dueñas<sup>11</sup>, Giovanna Zorzi<sup>2</sup>, Thomas Gasser<sup>1</sup>, Wassilios Meissner<sup>12</sup>, Anne-Catherine Bachoud-Lévi<sup>13</sup>, Bernhard Landwehrmeyer<sup>9</sup>, Juan Dario Ortigoza Escobar<sup>14</sup>, Ludger Schöls<sup>1</sup>, Holm Graessner<sup>1</sup>

<sup>1</sup>University Hospital Tuebingen, Germany; <sup>2</sup>Foundation IRCCS Neurological Institute Carlo Besta Milan, Italy; <sup>3</sup>Pediatric hospital Bambino Gesù, Rome, Italy; <sup>4</sup>Hospital Universitari Vall d'Hebron, Spain; <sup>5</sup>Assistance Publique-Hôpitaux de Paris, Hôpital Robert-Debré, France: Neuropédiatrie, CRMR LEUKOFRANCE, Paris, Reference centre for Leukodystrophies, France; <sup>6</sup>VU University Medical Center Amsterdam, Netherlands; <sup>7</sup>University Hospitals Leuven, Belgium; <sup>8</sup>Assistance Publique-Hôpitaux de Paris, Hôpital Pitié-Salpêtrière, France: Reference centre for rare dementias; <sup>9</sup>Universitätsklinikum Ulm, Germany; <sup>10</sup>Universitätsklinikum Schleswig-Holstein, Campus Luebeck, Germany; <sup>11</sup>Hospital Universitari Vall d'Hebron, Spain; <sup>12</sup>CRMR AMS, Service de Neurologie Bordeaux, France; <sup>13</sup>Assistance Publique-Hôpitaux de Paris, Hôpital Henri-Mondor, France: Reference centre for Huntington's disease; <sup>14</sup>Hospital Clínic i Provincial de Barcelona y Hospital de Sant Joan de Déu, Spain

## European Reference Network for Rare Neurological Diseases - ERN-RND

### Aim:

Faster diagnosis, appropriate treatment and care for RND patients

### Facts:

- Set-up in 2017, European initiative
- 31 Members & 10 Affiliated Partners in 22 EU countries, new Members to be included
- Members = European expertise centres for RND
- Patient centred

## Disease groups covered by ERN-RND

1. Ataxias and Hereditary Spastic Paraplegias
2. Atypical Parkinsonism and genetic Parkinson's disease
3. Dystonia, Paroxysmal Disorder and Neurodegeneration with Brain Iron Accumulation
4. Frontotemporal Dementia
5. Huntingtons' Disease and other Chorea
6. Leukodystrophies

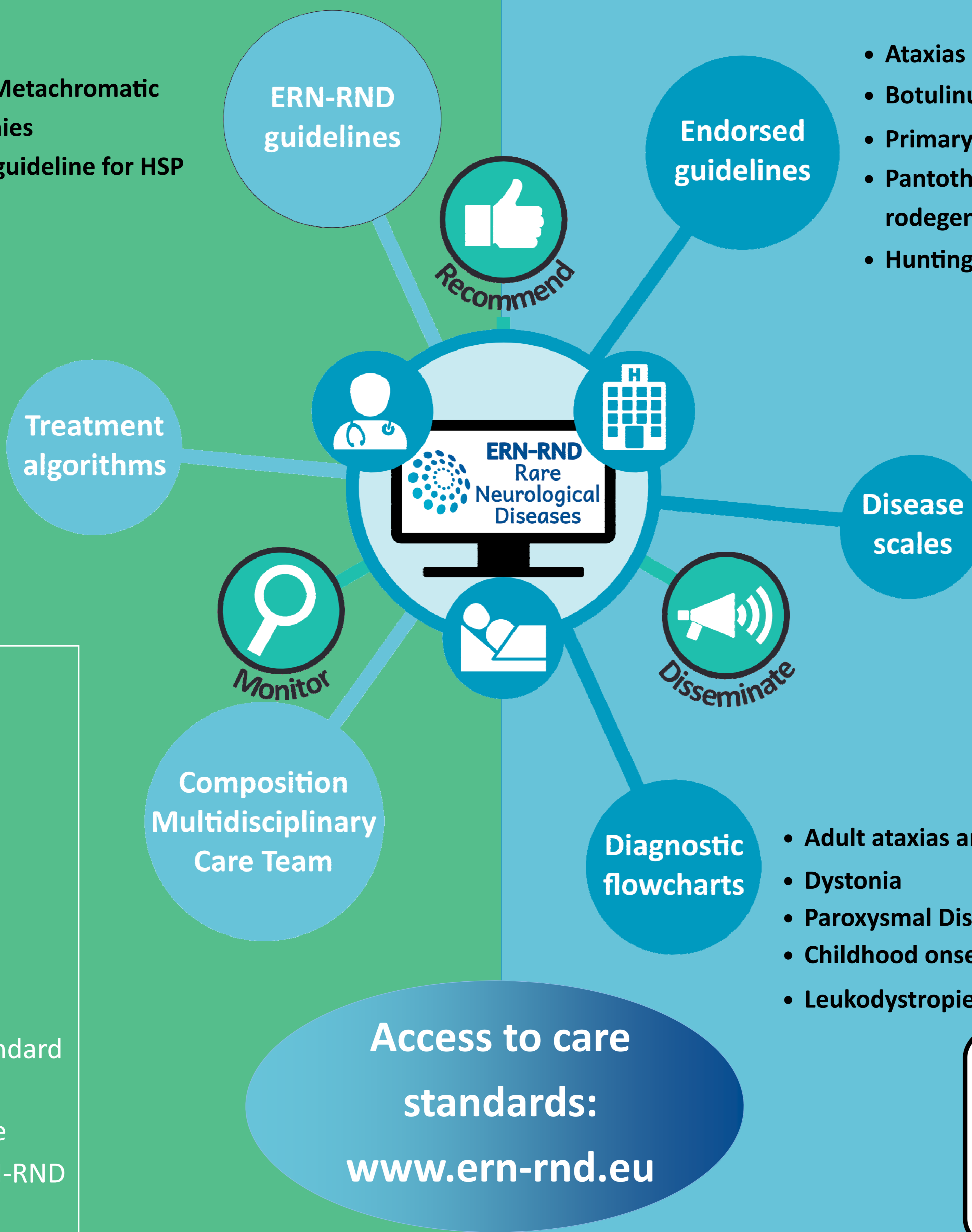
## UNDER DEVELOPMENT

- Guideline for Metachromatic Leukodystrophies
- Management guideline for HSP

- Dystonia
- Paroxysmal disorders

### ERN-RND roll-out concept for care-standards

1. Mapping of available information
2. Decision between endorsement and new development within disease group
3. Consent on care standard within disease group
4. Apply consented care standard across ERN-RND
5. Follow-up activities



Access to care standards:  
[www.ern-rnd.eu](http://www.ern-rnd.eu)

## IMPLEMENTED

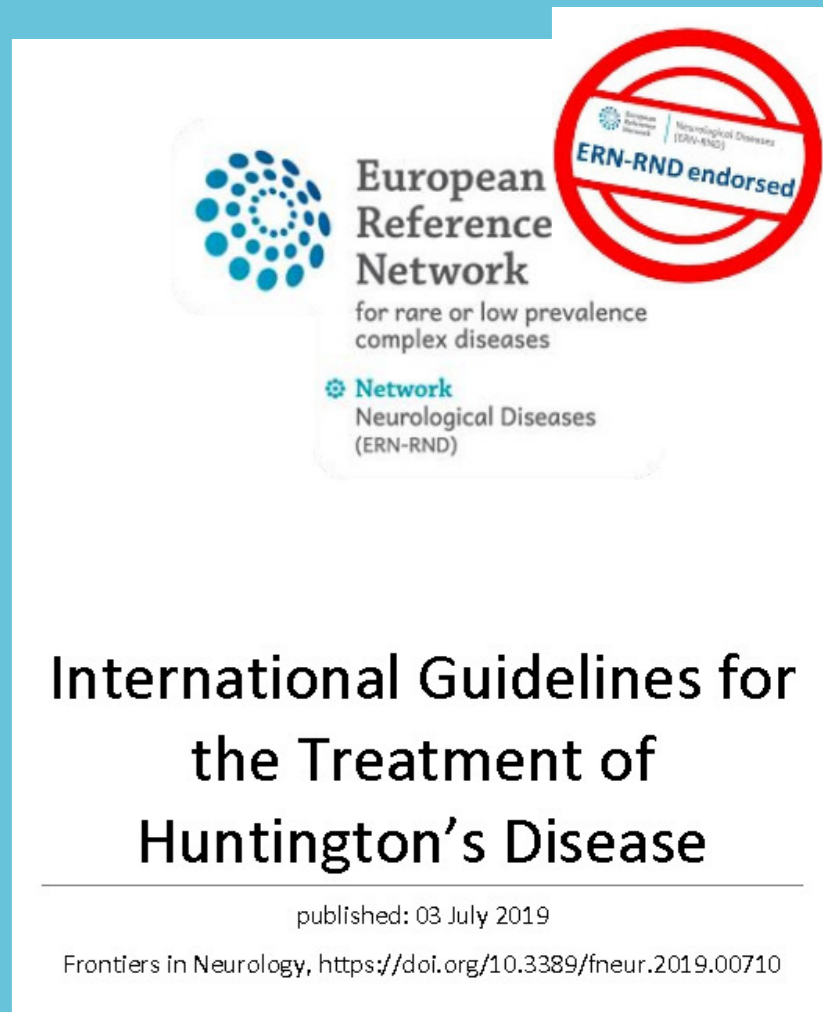
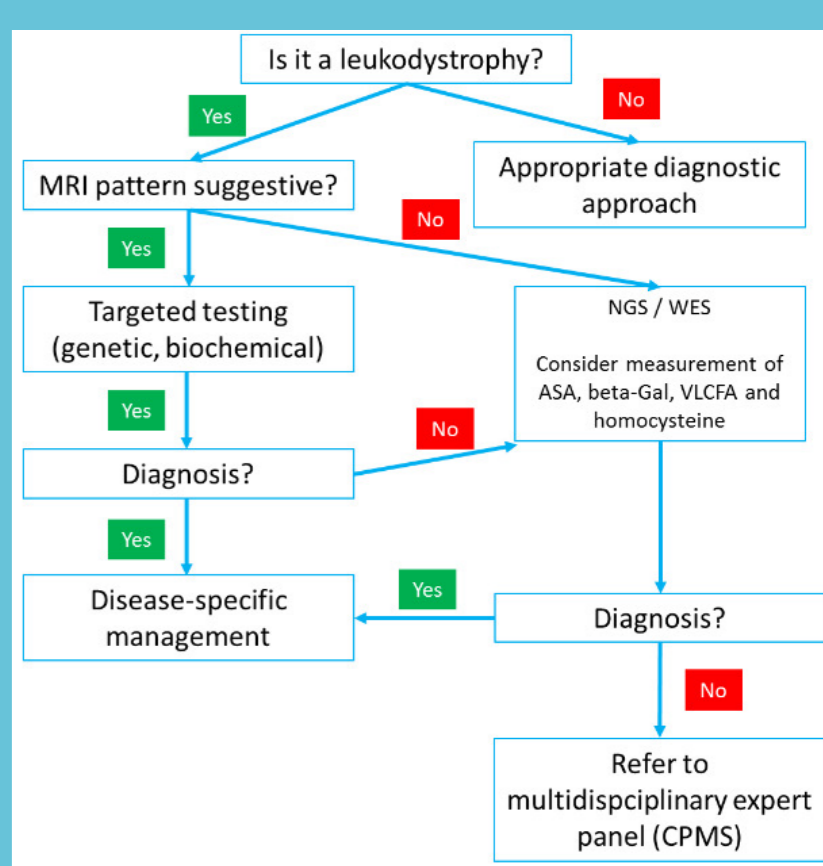
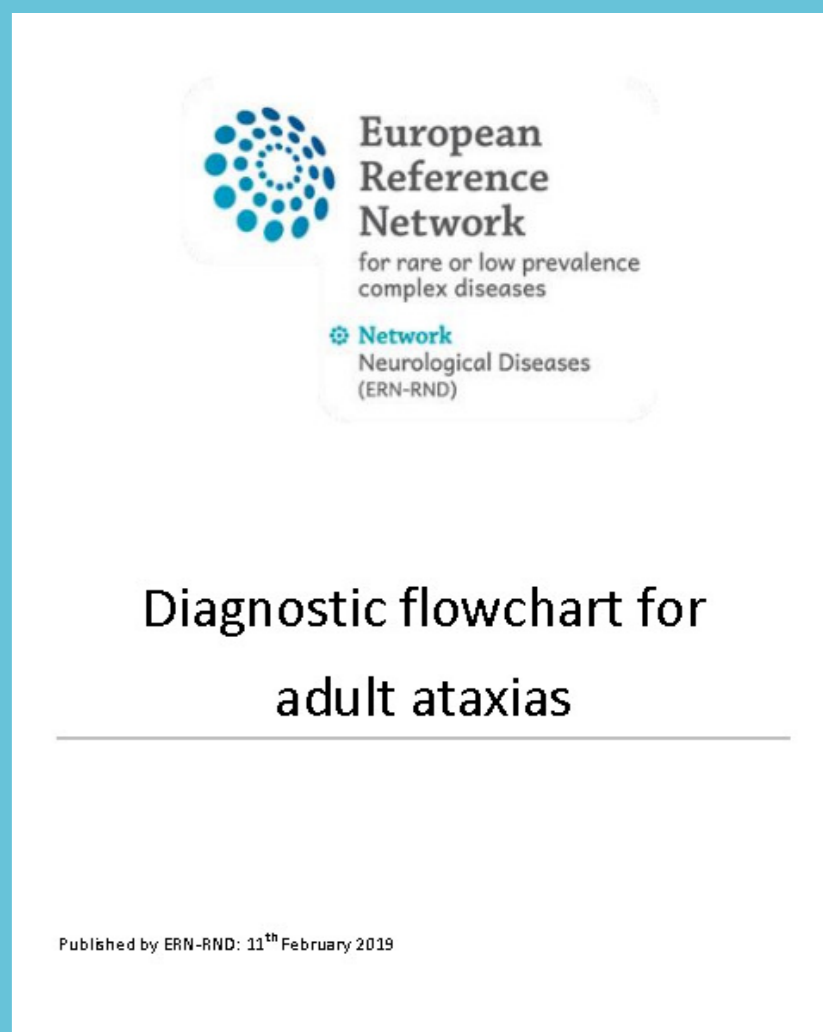
- Ataxias and Friedreich's ataxia
- Botulinum neurotoxin treatment
- Primary dystonias
- Pantothenate kinase-associated neurodegeneration (PKAN)
- Huntington's Disease

- Cerebellar Ataxia and HSP
- MSA, PSP and genetic PD
- Dystonia
- NBIA
- Chorea and HD

The different clinical rating scales are used by 20-95% of ERN-RND expert centers

- Adult ataxias and HSP
- Dystonia
- Paroxysmal Disorders
- Childhood onset Chorea
- Leukodystrophies

The different flow-charts are used by 45-85% of ERN-RND expert centers



Recommended scales:	Scale
Generalized dystonia	Scale for Motor Dyskinesia Rating Scale (SMR), Fahn's, Marsden CD, Brinman SS, Moskowitz C, Hershey W, and the reliability of a rating scale for the primary torsion dystonia. <i>Neurology</i> 1985;25:10-12
Generalized dystonia in children	The Movement Disorder-Childhood Rating Scale. Bertoni R, Spagnuolo G, Fattori E, Guzzetta A, Di Biase G, Scavone MC, Casali V, Marcolin L, Gini G. <i>Movement Disorders: Childhood Rating Scale, Reliability and Validity</i> . <i>Parkinsonism</i> 2008;24:204-205
Hemichorea	Chorea Rating Scale (CRS). Guzzetta A, Spagnuolo G, Fattori E, Bertoni R, Scavone MC, Casali V, Marcolin L, Gini G. <i>Movement Disorders: Childhood Rating Scale, Reliability and Validity</i> . <i>Parkinsonism</i> 2008;24:204-205
Cervical Dystonia	Toronto Western Spasmodic Torticollis Rating Scale for cervical dystonia - Conklin E, Brinman A, Fahn S, Marsden W, and Lang CE. <i>The Toronto Western Spasmodic Torticollis Rating Scale: Description, Assessment of validity and reliability</i> . <i>Neurology</i> 1991;40:485

