Diagnostic flowcharts
a tool to disseminate expert knowledge on rare neurological diseases

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ERN-RND roll-out concept for Diagnostic Flowcharts

- Mapping of existing diagnostic flowcharts and algorithms
- Consent on diagnostic flowchart for the different disease groups
- Apply consented diagnostic procedure across ERN-RND
- Follow-up activities needed for reaching consensus and possibly as basis for diagnostic guideline
- Proposal to all ERN-RND centres to implement flowcharts
- Dissemination of ERN-RND endorsed flowcharts to clinicians in Europe (website, newsletter, …)

ERN-RND Disease Groups

- Ataxias and Hereditary Spastic Paraplegias
- Atypical Parkinsonism and genetic PD
- Dystonia, Paroxysmal Disorder and Neurodegeneration with Brain Ion Accumulation
- Frontotemporal Dementia
- Huntington’s Disease and other Chorea
- Leukodystrophies

Diagnosis of patients with a rare neurological disorder

Step 1: Is it a dystonia?

Step 2: Causative factor identified, possibly other movement disorder

Step 3: Specific diagnostic work-up

Step 4: Perform recommended biochemical investigations (Table 1) and start levo-dopa trial

Step 5: Ned-generation sequencing (Supplement 1)

MRI pattern suggestive?

Appropriate diagnostic approach

Targeted testing (genetic, biochemical)

Diagnosis?

Disease-specific management

Refer to multidisciplinary expert panel (MDM)

ERN-RND develops and disseminates Diagnostic Flowcharts
to help guide the diagnosis of patients with a rare neurological disorder

The complete document lists a number of tests used at ERN-RND centres for PPA subtyping. We distinguish between tests that are typically used by the clinician during the clinical assessment and tests to be administered by a neuropsychologist or speech therapist during a more formal and extensive neurolinguistic or neuropsychological assessment. The purpose is to serve as a guide for neurologists and other specialties who are less experienced in PPA subtyping.