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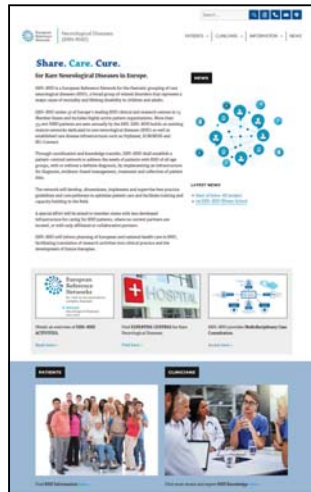
Key Facts and Figures

- ◆ 31 Healthcare Providers from 13 EU countries
- ◆ **Disease coverage:** Cerebellar Ataxias and Spastic Paraplegias, Chorea and Huntington's Disease, Dystonias, paroxysmal disorders (non-epileptical ones) and Neurodegeneration with Brain Iron Accumulation, Frontotemporal dementia, Leukodystrophies, Atypical parkinsonian syndromes: Genetic PD, Multisystem Atrophy, Progressive Supranuclear Palsy/Corticobasal degeneration
- ◆ Approximately 35.000 patients per year
 - 85% adult patients
 - 15% paediatric patients
 - Approximately 5.300 new patients per year
- ◆ Duration of first phase: five years (1.3.2017 - 28.02.2022)
- ◆ Patient centeredness through focus, approach and participation of patient representatives in all government and operational bodies

Knowledge generation and dissemination

To align quality of care provided in EU

- ◆ Expertise Centres
- ◆ Patient Organisations
- ◆ Disease Information
- ◆ Diagnostic algorithms
- ◆ Guidelines and consensus documents
- ◆ Published Care Evidence
- ◆ Clinical Rating Scales
- ◆ RND Registries
- ◆ Training and Education resources
- ◆ Disease Networks and Study Groups



Endorsement process: (1) PI from ERN-RND HCP to check for content, (2) Patient organisation to check for patient appropriateness

ERN-RND Network of Healthcare Providers

No.	Beneficiary	Country
1	Universitätsklinikum Tübingen	Germany
2	Université libre de Bruxelles	Belgium
3	General University Hospital in Prague	Czechia
4	CHU de Bordeaux	France
5	Assistance Publique-Hôpitaux de Paris, Hôpital Pitié-Salpêtrière	France
6	Universitätsklinikum Bonn	Germany
7	Universitätsklinikum Schleswig-Holstein	Germany
8	Klinikum der Universität München	Germany
9	University Hospitals Leuven	Belgium
10	Semmelweis University	Hungary
11	University of Pécs	Hungary
12	Foundation IRCCS neurological institute Carlo Besta - Milan	Italy
13	IRCCS Clinical Institute Humanitas - Rozzano - Milan	Italy
14	AOU Siena	Italy
15	Pediatric hospital Bambino Gesù, Rome	Italy
16	VU University Medical Center Amsterdam	Netherlands
17	University Medical Center Groningen	Netherlands
18	Radboud University Medical Center Nijmegen	Netherlands
19	University Medical Centre Ljubljana	Slovenia
20	Hospital Clínic i Provincial de Barcelona y Hospital de Sant Joan de Déu	Spain
21	Assistance Publique-Hôpitaux de Paris, Hôpital Henri-Mondor	France
22	CHU de Toulouse	France
23	Vilnius University Hospital Santariškių Klinikos	Lithuania
24	Hospital Universitari Vall d'Hebron - Barcelona	Spain
25	University College London Hospitals NHS Foundation Trust	United Kingdom
26	University Hospital in Krakow	Poland
27	Universitätsklinikum Ulm	Germany
28	University Neurological Hospital "St. Naum" Sofia	Bulgaria
29	Assistance Publique-Hôpitaux de Paris, Hôpital Robert-Debré	France
30	Motol University Hospital - Prague	Czechia
31	Erasmus MC: University Medical Center Rotterdam	Netherlands

Objectives

- ◆ To increase overall percentage of RND patients with a final diagnosis significantly
- ◆ To improve and harmonise care of RND patients across the EU
- ◆ To develop, share and implement care pathways and guidelines for all RND groups represented in ERN-RND
- ◆ To create, develop and enhance constituents' capacity to design, implement and supervise RND training, education and capacity building measures at the level of member states and of the network
- ◆ To develop a comprehensive and data based European RND cohort to better understand these conditions and thus improve their management and help developing and testing treatments
- ◆ To define minimum quality and interoperability criteria for RND registries

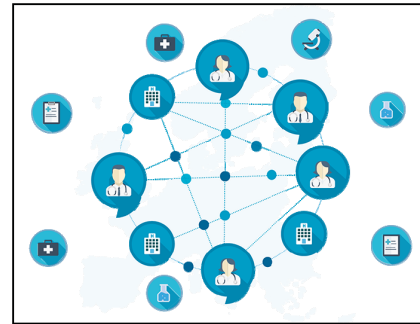
Clinical Patient Management System (CPMS)

The CPMS aims at supporting ERNs in improving the diagnosis and treatment of rare or low prevalence complex diseases across national borders of Member States in Europe.

Health professionals can use the CPMS to collaborate actively and share patient within and across ERNs. The CPMS is supported by Virtual Communication Tools and DICOM viewers to facilitate the interaction between clinicians.

ERN-RND will pursue three CPMS use cases in the next 12 months:

- ◆ Facilitate the role-out of the CPMS across the network through dedicated workshops and on-site visits to enable the network partners to discuss pilot cases.
- ◆ Make use of the platform to set-up a case based training functionality.
- ◆ Use the CPMS for identification and discussion of ultra-rare cases as planned in the Solve-RD project.



ERN-RND cohorts for Solve-RD

1. Unsolved cases

Definition: Unsolved RD cases with inconclusive exome; **Numbers:** at least 4,000 cases; **Main activities:** standardised collation of data & re-analysis with cutting-edge variant calling pipeline

2. Specific ERN cohorts

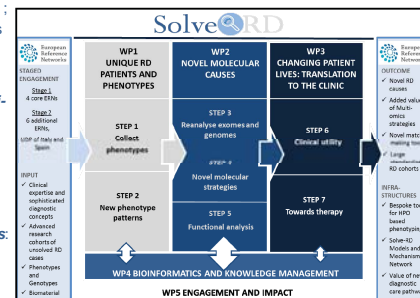
Definition: Disease group specific cohorts from; **Numbers:** 720 WGS (short reads), 100 WGS (long reads), 450 RNAseq (short reads), 60 RNAseq (long read); **Main activities:** „beyond the exome“ approaches

3. Ultra-rare Rare Diseases

Definition: Phenotypically unique RD patients/cohorts; **Numbers:** 300 cases; **Main activities:** Phenotype jamborees and exome/genome sequencing

4. The Unsolvable

Definition: Highly recognisable clinically defined diseases/syndromes for which no disease gene was identified yet (despite WES/WGS); **Numbers:** 20 cases; **Main activities:** Combination of all available omics tools to "crack" the "Unsolvable"



Key Targets of years 1 & 2

- ◆ Kick-off meeting
- ◆ Operability of all governance bodies as well as operational bodies
- ◆ Access points to all ERN-RND HCPs
- ◆ Introduction of CPMS in ERN-RND for e-consultation of clinical cases
- ◆ Consensus on disease group specific care quality indicators
- ◆ Consensus on diagnostic flowcharts for all RND covered by ERN-RND
- ◆ Identify disease groups specific most important care needs for RND in the EU
- ◆ Establish ERN-RND web-site as THE RND information hub