# The French national registry for rare diseases :
# A whole rare disease registry at national level meeting challenges at EU level

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**BNDMR**
Banque Nationale de Données
Maladies Rares



LOCAL

**1**

*Personal access*
Patients' monitoring
Local activity monitoring
Annual reporting

NATIONAL

**2**

*Analysis platform*
Epidemiological studies
Eligible patients identification for clinical trials
Care facilities need/offer (territorial breakdown, activity)
Mortality studies
*Medico-economic studies**
*Therapeutical observance**
*Patient care pathways**

Hospital EHR — *Connector* MDS — **BaMaRa** (1 per hospital) — MDS- De-identification — BNDMR Warehouse

INS

Health data national system

SNIIRAM
PMSI
CépiDc
...

*requieres a unique national identifier (INS)*

## Context and objectives

The French national registry for rare diseases (BNDMR) was created as part of the second National Plan for Rare Diseases. Its aim is to collect from all expert centers on rare diseases in France a Minimal Data Set (MDS), either from a web app (BaMaRa) or from the electronic health record (EHR). In the end, these data are sent to the BNDMR data warehouse to allow national analyses.

The BNDMR team is also committed to directly make possible the upload of its data to the forthcoming ERN registries and CPMS platform to avoid multiple inputs for the same data for professionals.
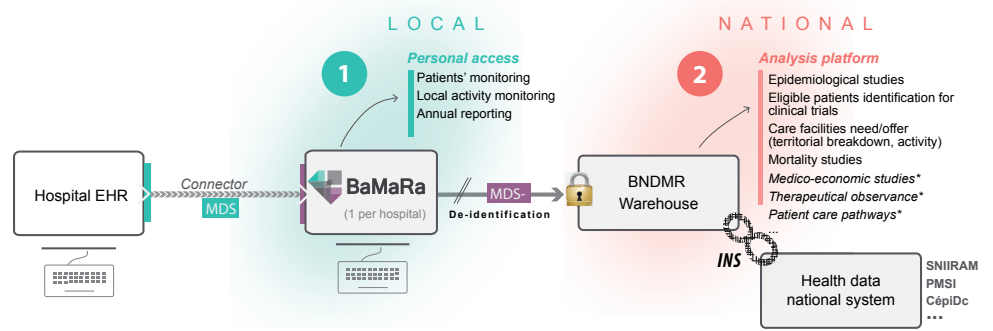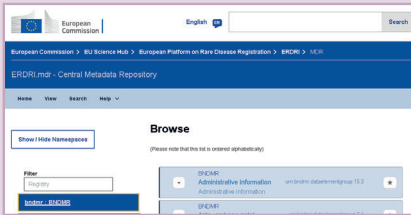
## Results (March 2020)

### The RD French minimum data set (MDS)

The French MDS contains about 60 items, including :

- Patient identification
- Vital status
- Care pathway & activities
- Ante and neonatal data
- Treatment (orphan drugs)
- Diagnosis (ORPHAcodes)
- Diagnostic history and accuracy
- Phenotype (HPO / ICD-10)
- Genotype (HGNC / HGVS)

Download it on our website: http://www.bndmr.fr/le-set-de-donnees-minimal/
Items also listed on the European Commission ERDRI.mdr platform:
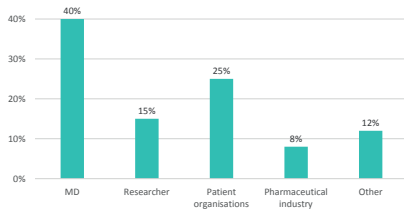https://eu-rd-platform.jrc.ec.europa.eu/mdr/view.xhtml

It was the source of the Common data elements of the European Commission constituting the core of ERN registries, therefore it natively overlaps with it.
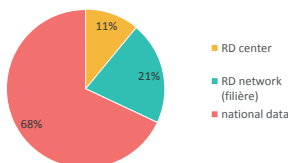
### Study requests made on the BNDMR data

**30 applications** received in **2019**
**+ 12** during the first trimester **2020**

#### Various stakeholders ask for BNDMR data

| Stakeholder | Percentage |
| --- | --- |
| MD | 40% |
| Researcher | 15% |
| Patient organisations | 25% |
| Pharmaceutical industry | 8% |
| Other | 12% |

#### Granularity level of the data requests

- 11% RD center
- 21% RD network (filière)
- 68% national data

### 1 The participating network

**2 700 professionnals** involves in the data collection *(medical or paramedical staff, secretary, clinical research associate...)*

**67 hospitals** including overseas

**820 expert centres** actively collecting data

### 2 The database content

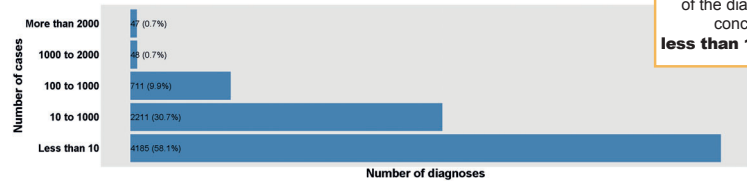**700 000 patients** (3,2% declared as deceased)
**22 700 healthy carriers**

Incl. **37 500 foetuses** — 33% deceased

### 3 The rare diseases diagnosis

**4 640 diagnostic terms** (Orphanet omenclature)

#### Distribution of diagnoses by number of cases (in classes)

| Number of cases | |
| --- | --- |
| More than 2000 | 47 (0.7%) |
| 1000 to 2000 | 48 (0.7%) |
| 100 to 1000 | 711 (9.9%) |
| 10 to 1000 | 2211 (30.7%) |
| Less than 10 | 4185 (58.1%) |

Number of diagnoses

**More than half** of the diagnoses concern **less than 10 cases**

#### Level of diagnostic confirmation

| | |
| --- | --- |
| Undetermined | 258903 (26.5%) |
| Ongoing | 95493 (9.8%) |
| Likely | 91885 (9.4%) |
| Confirmed | 529415 (54.3%) |

Number of cases

**Confirmed diagnosis** in only **1 out of 2 cases**