

# French National Registry of Rare Diseases (Banque Nationale de Données Maladies Rares)

**First published:** 28/09/2022

**Last updated:** 17/10/2024

Data source

Human

Disease registry

Other

## Administrative details

### Administrative details

#### Data source ID

1111176

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#### Data source acronym

BNDMR

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#### Data holder

[Assistance Publique - Hôpitaux de Paris \(AP-HP\)](#)

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#### Data source type

Disease registry

Other

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## Data source type, other

National rare diseases registry

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## Main financial support

Funding from public-private partnership  
National, regional, or municipal public funding

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## Care setting

Secondary care – specialist level (ambulatory)

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## Data source qualification

If the data source has successfully undergone a formal qualification process (e.g., from the EMA, ISO or other certifications), this should be described.

Yes

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## Description of the qualification

French Health Authority (HAS)

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## Data source website

<https://www.bndmr.fr/>

## Contact details

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## Data source regions and languages

## Data source countries

France

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## Data source languages

English

French

## Data source establishment

### Data source established

15/06/2017

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### Data source time span

**First collection:** 15/06/2017

The date when data started to be collected or extracted.

## Publications

### Data source publications

[The ongoing French BaMaRa-BNDMR cohort: implementation and deployment of a nationwide information system on rare disease](#)

[French data on the epidemiology and expert healthcare network for epidermolysis bullosa](#)

[Prevalence of fibrodysplasia ossificans progressiva \(FOP\) in France: an estimate based on a record linkage of two national databases](#)

[10 years of CEMARA database in the AnDDI-Rares network: a unique resource facilitating research and epidemiology in developmental disorders in France](#)

[Impact of the COVID-19 pandemic on the care of rare and undiagnosed diseases patients in France: a longitudinal population-based study](#)

## Data elements collected

## **Disease information**

Does the data source collect information with a focus on a specific disease? This might be a patient registry or other similar initiatives.

Yes

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## **Disease details (other)**

Rare disease common data elements for all patients attending rare disease expert centers (Rémy Choquet, Meriem Maaroufi, Albane de Carrara, Claude Messiaen, Emmanuel Luigi, Paul Landais. A methodology for a minimum data set for rare diseases to support national centers of excellence for healthcare and research. J Am Med Inform Assoc. 2015 Jan;22(1):76-85.)

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## **Rare diseases**

Are rare diseases captured? In the European Union a rare disease is one that affects no more than 5 people in 10,000.

Yes

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## **Pregnancy and/or neonates**

Does the data source collect information on pregnant women and/or neonatal subpopulation (under 28 days of age)?

Yes

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## **Hospital admission and/or discharge**

Yes

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## **ICU admission**

Is information on intensive care unit admission available?

No

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## **Cause of death**

Captured

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**Cause of death vocabulary**

ICD-10

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**Prescriptions of medicines**

Captured

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**Prescriptions vocabulary**

other

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**Dispensing of medicines**

Not Captured

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**Advanced therapy medicinal products (ATMP)**

Is information on advanced therapy medicinal products included? A medicinal product for human use that is either a gene therapy medicinal product, a somatic cell therapy product or a tissue engineered products as defined in Regulation (EC) No 1394/2007 [Reg (EC) No 1394/2007 Art 1(1)].

Yes

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**Contraception**

Is information on the use of any type of contraception (oral, injectable, devices etc.) available?

No

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**Indication for use**

Does the data source capture information on the therapeutic indication for the use of medicinal products?

Captured

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**Indication vocabulary**

Other

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## **Indication vocabulary, other**

HPO, ORPHAcodes, ICD10 HGNC (if gene therapy)

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## **Medical devices**

Is information on medicinal devices (e.g., pens, syringes, inhalers) available?

No

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## **Administration of vaccines**

No

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## **Procedures**

Does the data source capture information on procedures (e.g., diagnostic tests, therapeutic, surgical interventions)?

Captured

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## **Procedures vocabulary**

Not coded (Free text)

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## **Healthcare provider**

Is information on the person providing healthcare (e.g., physician, pharmacist, specialist) available?  
The healthcare provider refers to individual health professionals or a health facility organisation licensed to provide health care diagnosis and treatment services including medication, surgery and medical devices.

Yes

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## **Clinical measurements**

Is information on clinical measurements (e.g., BMI, blood pressure, height) available?

No

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## **Genetic data**

Are data related to genotyping, genome sequencing available?

Captured

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### **Genetic data vocabulary**

HGNC

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### **Biomarker data**

Does the data source capture biomarker information? The term “biomarker” refers to a broad subcategory of medical signs ( objective indications of medical state observed from outside the patient), which can be measured accurately and reproducibly. For example, haematological assays, infectious disease markers or metabolomic biomarkers.

Captured

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### **Biomarker data vocabulary**

Other

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### **Biomarker vocabulary, other**

HPO

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### **Patient-reported outcomes**

Is information on patient-reported outcomes (e.g., quality of life) available?

No

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### **Patient-generated data**

Is patient-generated information (e.g., from wearable devices) available?

No

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### **Units of healthcare utilisation**

Are units of healthcare utilisation (e.g., number of visits to GP per year, number of hospital days) available or can they be derived? Units of healthcare utilisation refer to the quantification of the use of services for the purpose of preventing or curing health problems.

Yes

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**Unique identifier for persons**

Are patients uniquely identified in the data source?

Yes

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**Diagnostic codes**

Captured

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**Diagnosis / medical event vocabulary**

Other

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**Diagnosis / medical event vocabulary, other**

ORPHAcodes, HPO

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**Medicinal product information**

Captured

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**Medicinal product information collected**

Active ingredient(s)

Brand name

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**Medicinal product vocabulary**

ART 57

Other

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**If 'other,' what vocabulary is used?**

EMA Orphan drugs registry, Theriaque

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**Quality of life measurements**

Not Captured

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## **Lifestyle factors**

Not Captured

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## **Sociodemographic information**

Captured

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## **Sociodemographic information collected**

Age

Country of origin

Gender

Other

Sex

Type of residency

## Quantitative descriptors

## Population Qualitative Data

### **Population age groups**

Paediatric Population (< 18 years)

Preterm newborn infants (0 - 27 days)

Term newborn infants (0 - 27 days)

Infants and toddlers (28 days - 23 months)

Children (2 to < 12 years)

Adolescents (12 to < 18 years)

Adults (18 to < 46 years)

Adults (46 to < 65 years)

Elderly ( $\geq 65$  years)

Adults (65 to  $< 75$  years)

Adults (75 to  $< 85$  years)

Adults (85 years and over)

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**Estimated percentage of the population covered by the data source in the catchment area**

99% of patients visiting rare disease experts' centres

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**Description of the population covered by the data source in the catchment area whose data are not collected (e.g., people who are registered only for private care)**

All patients suffering from rare diseases and visiting rare disease expert centres

## Family linkage

**Family linkage available in the data source permanently or can be created on an ad hoc basis**

Ad hoc

## Population

**Population size**

1000000

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**Active population size**

1185441

## Population by age group

Age group	Population size	Active population size
Paediatric Population (< 18 years)	479704	469663
Term newborn infants (0 - 27 days)	12193	10413
Infants and toddlers (28 days - 23 months)	99225	94648
Children (2 to < 12 years)	242280	239803
Adolescents (12 to < 18 years)	126006	124799
Adults (18 to < 46 years)	398745	393805
Adults (46 to < 65 years)	204058	194398
Elderly ( $\geq$ 65 years)	148378	127575
Adults (65 to < 75 years)	87702	78370
Adults (75 to < 85 years)	47899	39896
Adults (85 years and over)	12777	9309

## Median observation time

**Median time (years) between first and last available records for unique individuals captured in the data source**

0.84

**Median time (years) between first and last available records for unique active individuals (alive and currently registered) captured**

1.00

## **Governance details**

Documents or webpages that describe the overall governance of the data source and processes and procedures for data capture and management, data quality check and validation results (governing data access or utilisation for research purposes).

<https://www.bndmr.fr/exploiter/donnees-bndmr/charte-bndmr/>

<https://www.bndmr.fr/le-projet/la-gouvernance/>

[https://www.bndmr.fr/wp-content/uploads/2019/10/COSCI\\_BNDMR\\_REGLEMENT-INTERIEUR.pdf](https://www.bndmr.fr/wp-content/uploads/2019/10/COSCI_BNDMR_REGLEMENT-INTERIEUR.pdf)

## **Biospecimen access**

Are biospecimens available in the data source (e.g., tissue samples)?

No

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## **Access to subject details**

Can individual patients/practitioners/practices included in the data source be contacted?

No

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## **Description of data collection**

Data collected by health provider through EHR or BaMaRa application directly.  
Data are then pseudonymized into the BNDMR.

# Event triggering registration

## **Event triggering registration of a person in the data source**

Disease diagnosis

Practice registration

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## **Event triggering de-registration of a person in the data source**

Other

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**Event triggering de-registration of a person in the data source, other**  
objection to the re-use of their data for research

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**Event triggering creation of a record in the data source**  
First visit in the rare disease expert centre

## Data source linkage

### Linkage

Is the data source described created by the linkage of other data sources (prelinked data source) and/or can the data source be linked to other data source on an ad-hoc basis?

Yes

## Linked data sources

### Pre linked

Is the data source described created by the linkage of other data sources?

No

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### Data source, other

French Health Data System

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### Linkage strategy

Deterministic

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### Linkage variable

National identifier for deterministic linkage. Date of birth, sex, health care providers and outpatient and inpatient visit dates.

### **Pre linked**

Is the data source described created by the linkage of other data sources?

Yes

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### **Data source, other**

French National Registry of deceased persons

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### **Linkage strategy**

Combination

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### **Linkage variable**

Name, Surname, Birthdate, Sex

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### **Linkage completeness**

85.00%

## Data management specifications that apply for the data source

### **Data source refresh**

Monthly

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### **Informed consent for use of data for research**

Required for intervention studies

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### **Possibility of data validation**

Can validity of the data in the data source be verified (e.g., access to original medical charts)?

Yes

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### **Data source preservation**

Are records preserved in the data source indefinitely?

No

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### **Data source preservation length (years)**

20 years

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### **Approval for publication**

Is an approval needed for publishing the results of a study using the data source?

No

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### **Data source last refresh**

01/04/2023

## **Common Data Model (CDM) mapping**

### **CDM mapping**

Has the data source been converted (ETL-ed) to a common data model?

Yes

### **CDM Mappings**

#### **CDM name (other)**

SDM-MR

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#### **Data source ETL specifications (link)**

<https://www.bndmr.fr/boite-a-outils/kit-editeurs/sdm-mr-cda/>