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

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PURI

https://redirect.ema.europa.eu/resource/1111176

Data source ID

1111176

Data source acronym

BNDMR

Data holder

Assistance Publique - Hôpitaux de Paris (AP-HP)

Data source type

Disease registry

Other

Data source type, other

National rare diseases registry

Main financial support

Funding from public-private partnership

National, regional, or municipal public funding

Care setting

Secondary care – specialist level (ambulatory)

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

Description of the qualification

French Health Authority (HAS)

Data source website

https://www.bndmr.fr/

Contact details

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Data source regions and languages

Data source countries

France

Data source languages

English

French

Data source establishment

Data source established

15/06/2017

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

The data source contains the following information

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

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.)

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

Pregnancy and/or neonates

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

Yes

Hospital admission and/or discharge

Yes

ICU admission

Is information on intensive care unit admission available?

No

Cause of death

Captured

Cause of death vocabulary

ICD-10

Prescriptions of medicines

Captured

Prescriptions vocabulary

other

Dispensing of medicines

Not Captured

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

Contraception

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

No

Indication for use

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

Captured

Indication vocabulary

Other

Indication vocabulary, other

HPO, ORPHAcodes, ICD10 HGNC (if gene therapy)

Medical devices

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

No

Administration of vaccines

No

Procedures

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

Captured

Procedures vocabulary

Not coded (Free text)

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

Clinical measurements

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

No

Genetic data

Are data related to genotyping, genome sequencing available?

Captured

Genetic data vocabulary

HGNC

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

Biomarker data vocabulary

Other

Biomarker vocabulary, other

HPO

Patient-reported outcomes

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

No

Patient-generated data

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

No

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

Unique identifier for persons

Are patients uniquely identified in the data source?

Yes

Diagnostic codes

Captured

Diagnosis / medical event vocabulary

Other

Diagnosis / medical event vocabulary, other

ORPHAcodes, HPO

Medicinal product information

Captured

Medicinal product information collected

Active ingredient(s)

Brand name

Medicinal product vocabulary

ART 57

Other

If 'other,' what vocabulary is used?

EMA Orphan drugs registry, Theriaque

Quality of life measurements

Not Captured

Lifestyle factors

Not Captured

Sociodemographic information

Captured

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 (≥ 65 years)

Adults (65 to < 75 years)

Adults (75 to < 85 years)

Adults (85 years and over)

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

99% of patients visiting rare disease experts’ centres

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

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 (≥ 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) capt

1.00

Data flows and management

Access and validation

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

Biospecimen access

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

No

Access to subject details

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

No

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

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

Other

Event triggering de-registration of a person in the data source, other

objection to the re-use of their data for research

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

Data source, other

French Health Data System

Linkage strategy

Deterministic

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

Data source, other

French National Registry of deceased persons

Linkage strategy

Combination

Linkage variable

Name, Surname, Birthdate, Sex

Linkage completeness

85.00%

Data management specifications that apply for the data source

Data source refresh

Monthly

Informed consent for use of data for research

Required for intervention studies

Possibility of data validation

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

Yes

Data source preservation

Are records preserved in the data source indefinitely?

No

Data source preservation length (years)

20 years

Approval for publication

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

No

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

Data source ETL specifications (link)

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