Gathering in a single database one of the largest rare disease cohort in the world: **the French national rare disease registry data**

BNDMR
Banque Nationale de Données Maladies Rares
bndmr.fr

Thibaut Pichon, Claude Messiaen, Louis Soussand, Céline Angin, Nabila Elarouci, Arnaud Sandrin, Anne-Sophie Jannot
Banque Nationale de Données Maladies Rares – BNDMR (AP-HP), Paris (France) contact.bndmr@aphp.fr

Context

French National Rare Disease Registry (Banque Nationale de Données Maladies Rares, BNDMR) collects data from all patients attending rare disease expert centre, with a nearly complete coverage since 2021.

Objectives

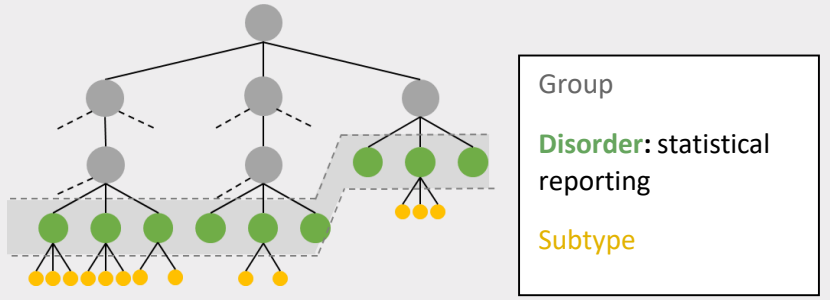
Estimate the number of recorded unique patients per rare disease in this data collection and compare it with published case reports and reported prevalence and incidence.

Population and methods

- Count of the number of patients for each rare disease after deduplication across hospitals and after excluding foetuses and foreign residents.
- For each rare disease, we extracted Orphanet data on published case reports and reported prevalence and incidence.

Rare diseases coding in the BNDMR

- To identify the patients' rare disease(s), the BNDMR uses the Orphanet nomenclature (July 2021 version).
- Each clinical entity is assigned a unique identifier that is stable over time: the ORPHAcode.
- ORPHAcodes are organised into three classification levels: disease groups, diseases and disease subtypes.



Orphanet nomenclature classification levels representation

Estimation of the number of recorded unique patients per rare disease in the BNDMR

Patients attending rare disease expert centres

Exclusion of foetuses and foreign residents

Exclusion of patients based on altered diagnosis coding

Patients without a diagnosis ORPHAcode in the database (n= 200 000)
Patients with diagnosis ORPHAcode now considered as not rare in Europe (n= 53 000)
Patients with group level ORPHAcode

Aggregation of Subtypes at corresponding Disorder level

Deduplication across hospitals

Count by rare diseases

2092 different rare diseases with more than 10 patients

490 595 patients

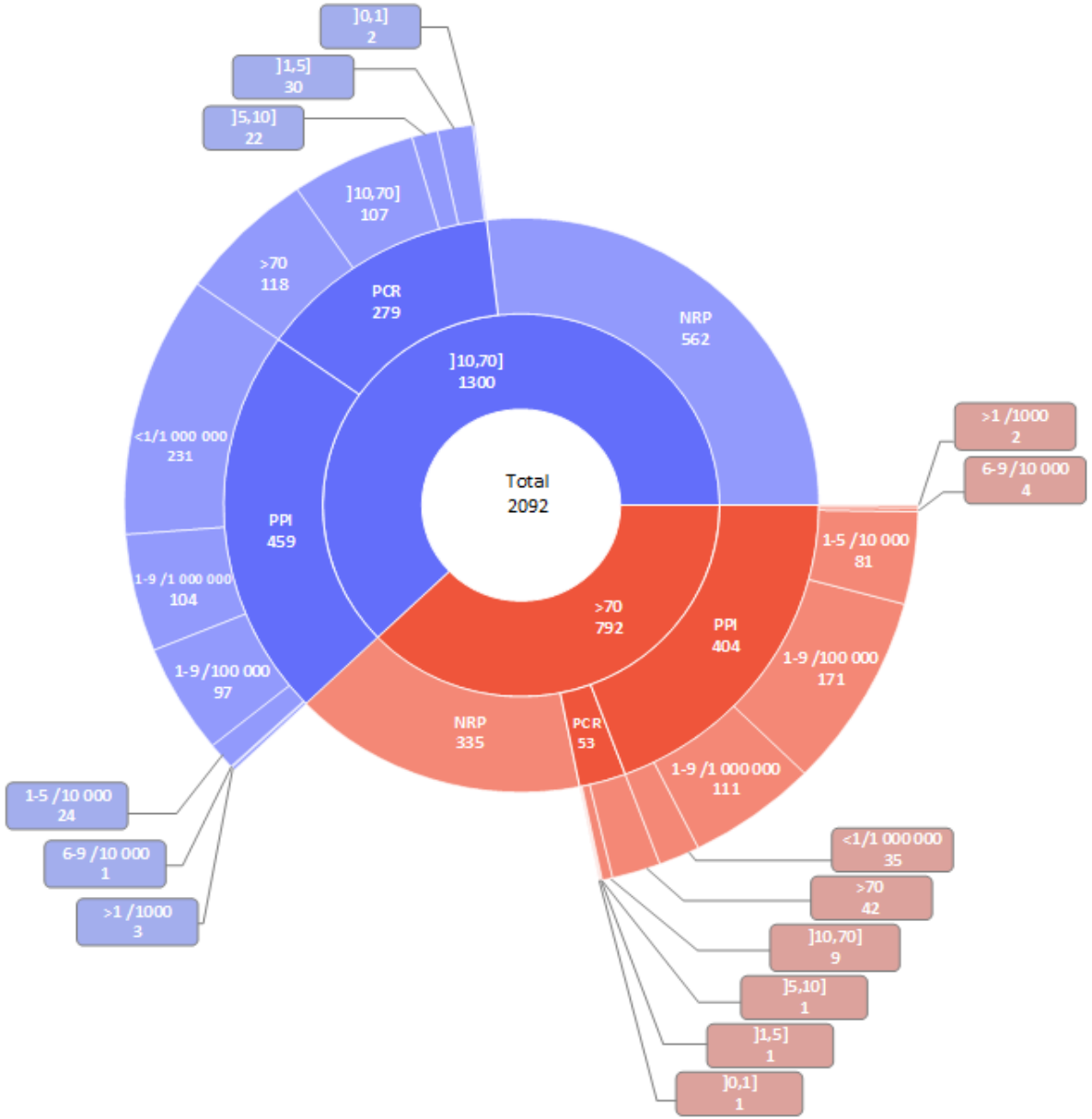
Open access available at

https://www.bndmr.fr/publications/nombre-de-cas-par-mr/

BNDMR
Banque Nationale de Données Maladies Rares

ASSISTANCE PUBLIQUE HÔPITAUX DE PARIS

Comparison with published case reports and reported prevalence and incidence



Number of diagnoses according to cohort size (darker shading). For each cohort size class, rare disease diagnoses are further distributed according to available Orphanet data (medium shading). These subclasses are further divided according to point prevalence or incidence estimates or number of disease cases reported by Orphanet (lighter shading).

***NRP** = no publication reported by Orphanet,*
***PPI**= point prevalence or incidence publication(s) reported by Orphanet,*
***PCR**= case report(s) reported by Orphanet.*

The BNDMR can be a valuable source for clinical research to enhance patients' recruitment in clinical trials and to better understand rare disease clinical pictures and epidemiology, by gathering in a single registry one of the largest RD cohort in the world.

BNDMR
Banque Nationale de Données Maladies Rares

ASSISTANCE PUBLIQUE HÔPITAUX DE PARIS