Orphanet, reference source of epidemiological data on rare diseases

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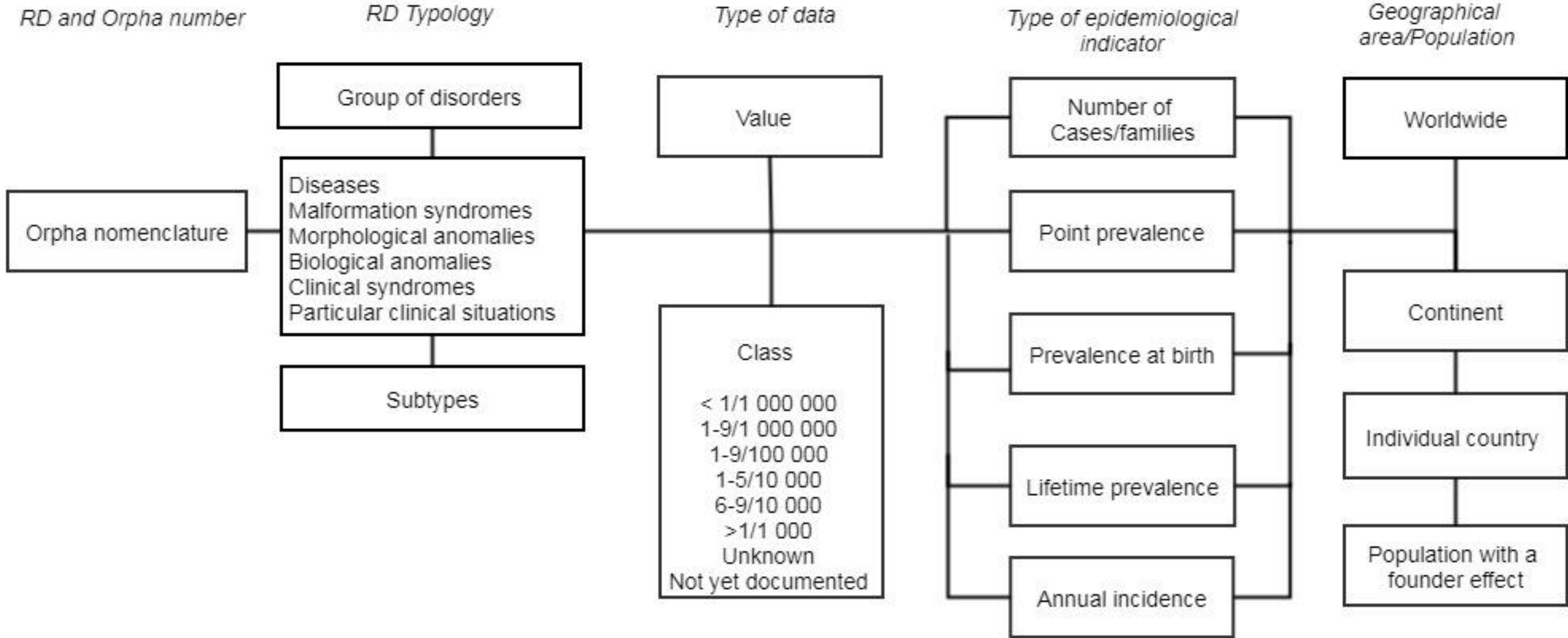INSERM, US14-Orphanet, Plateforme Maladies Rares, 96 rue Didot, 75014, Paris, France

Introduction

Despite the major advances in understanding rare diseases (RD) in recent years, there remains gaps in terms of epidemiological data concerning these diseases, as population-based studies and registries are few and far between. Orphanet (www.orpha.net), the European RD knowledge base, is developing strategies to improve epidemiological information on RD.
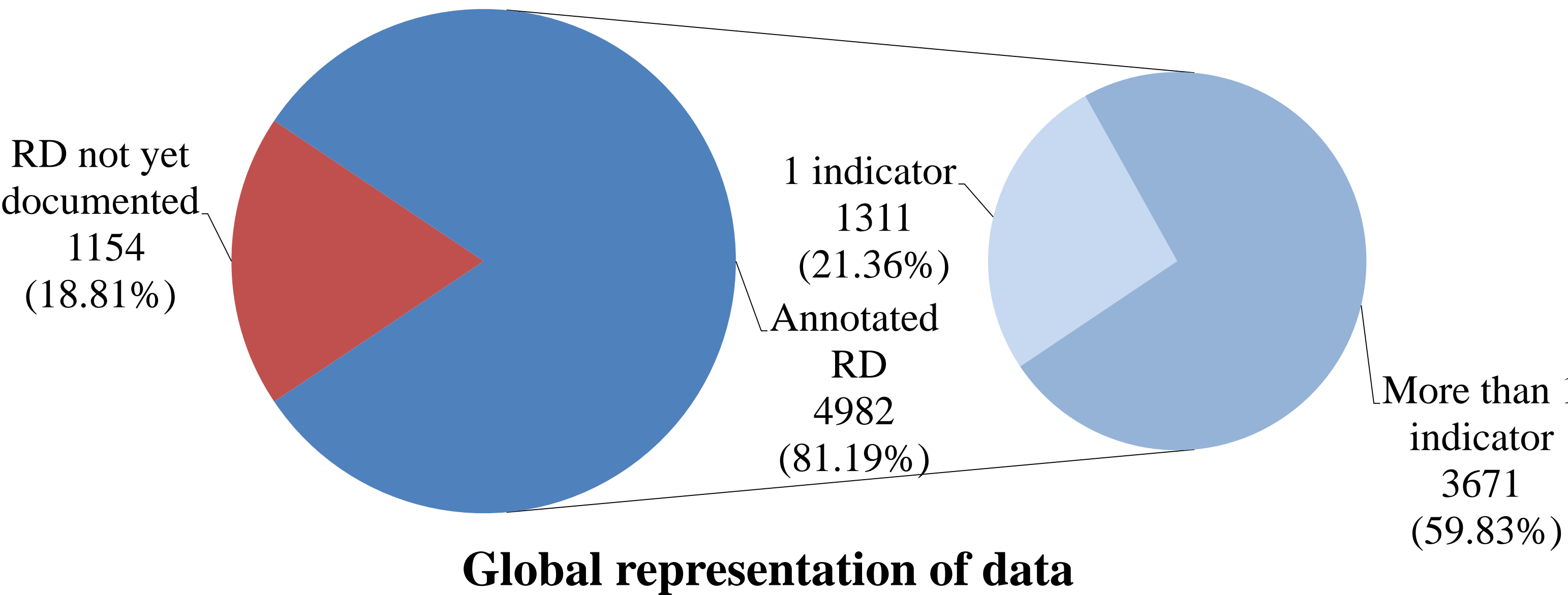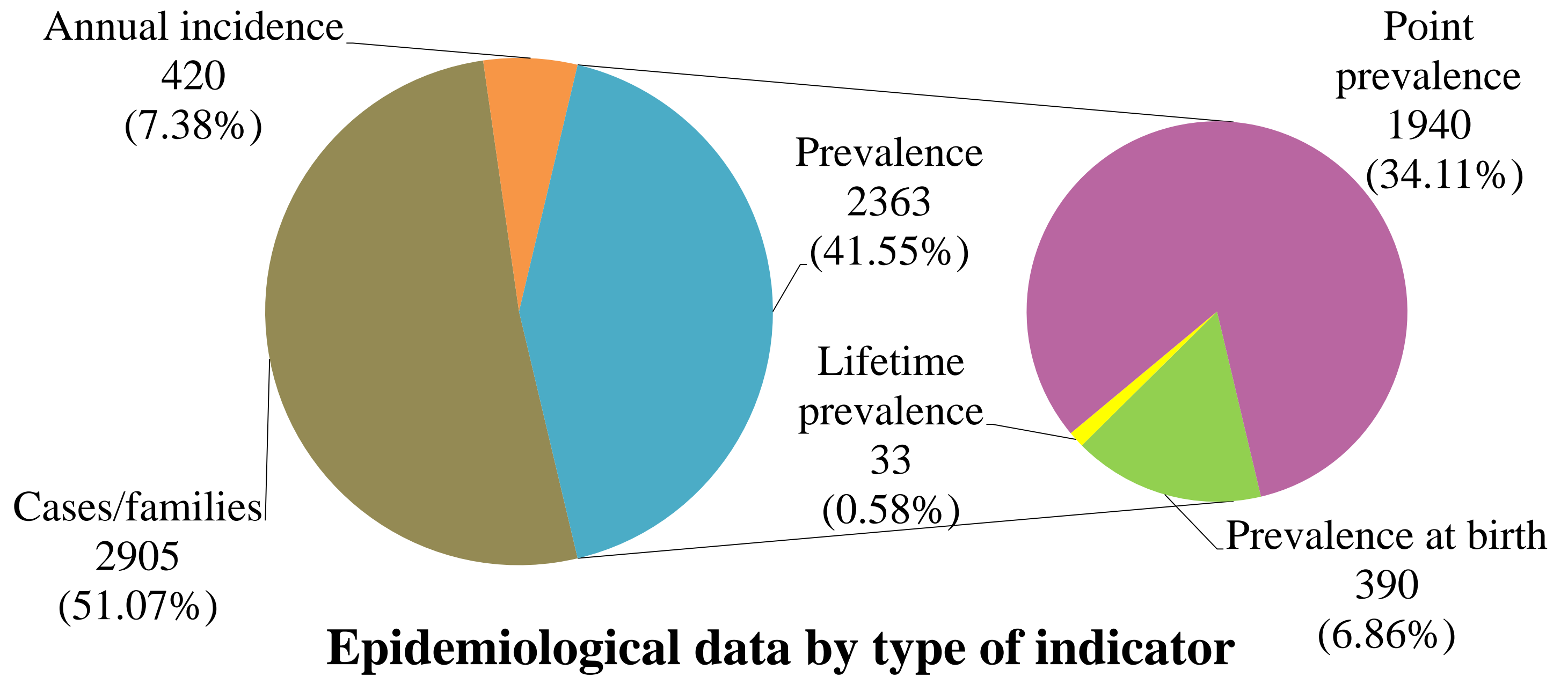
Methods

Epidemiological data life cycle

- 1. Data collection
  - Literature survey
  - Orphan designation
  - Registries
  - Networks
- 2. Analysis
  - Averages
  - Geographical extrapolation
- 3. Validation
  - Expert advice
  - Orphanet quality criteria
- 4. Quality control
  - Consistency
  - Comprehensiveness

Representation of Epidemiological data in the Orphanet database

Results

Global representation of data

- RD not yet documented 1154 (18.81%)
- Annotated RD 4982 (81.19%)
- 1 indicator 1311 (21.36%)
- More than 1 indicator 3671 (59.83%)

Epidemiological data by type of indicator

- Annual incidence 420 (7.38%)
- Prevalence 2363 (41.55%)
- Cases/families 2905 (51.07%)
- Lifetime prevalence 33 (0.58%)
- Point prevalence 1940 (34.11%)
- Prevalence at birth 390 (6.86%)
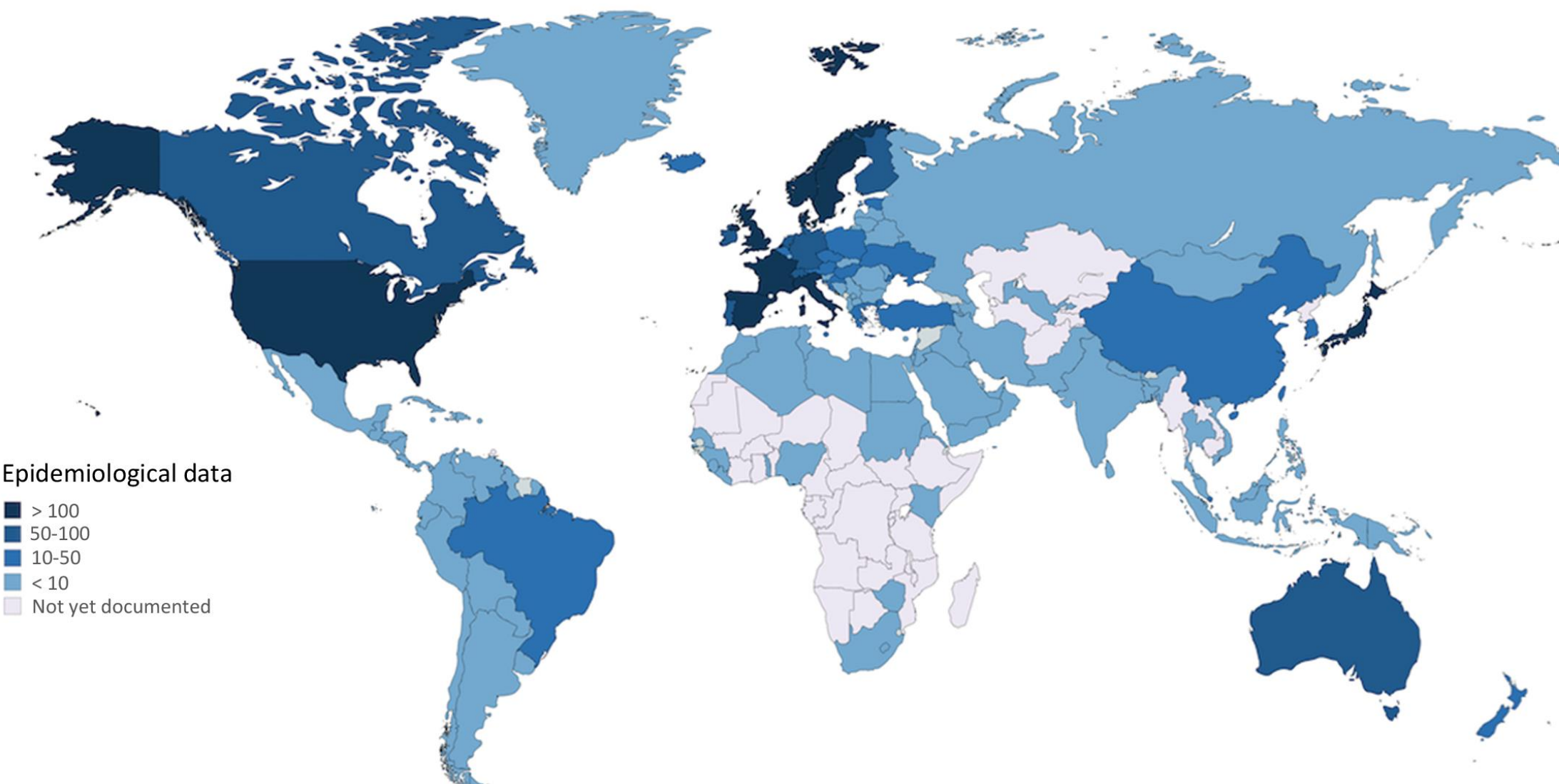
Geographic coverage of epidemiological data
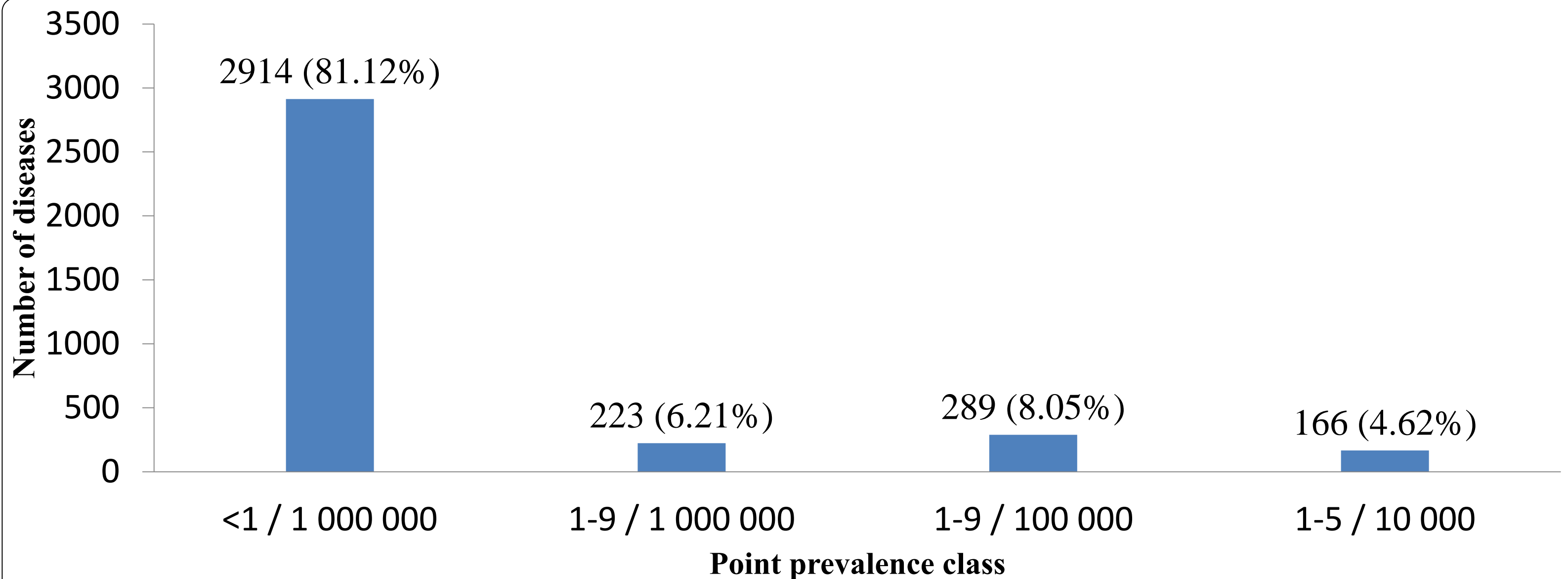
Point prevalence class distribution

- <1 / 1 000 000 : 2914 (81.12%)
- 1-9 / 1 000 000 : 223 (6.21%)
- 1-9 / 100 000 : 289 (8.05%)
- 1-5 / 10 000 : 166 (4.62%)

Conclusion

With its wide coverage of epidemiological data about RD, Orphanet represents the most comprehensive source of validated RD epidemiological data. This is of high interest for political decision-makers, researchers and the pharmaceutical industry. Analysis of this data will allow more accurate estimates of the global prevalence of RD in the general population to be carried out.

Information on a specific disorder
www.orpha.net

Data compilation
Worldwide and Europe
(Published biannually)

Computational use subset of disorders/ data under DTA/service contract
www.orphadata.org
(Updated montly)