

Orphanet, reference source of epidemiological data on rare diseases

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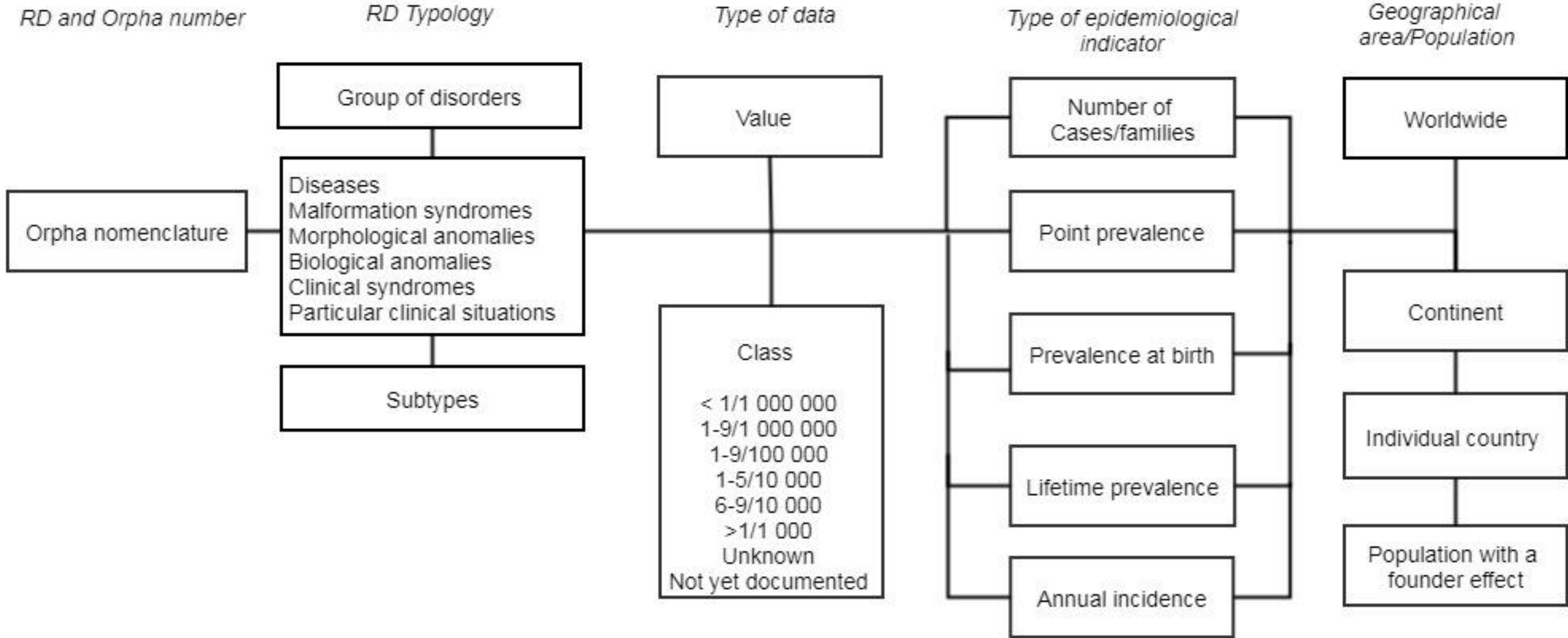
Introduction

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

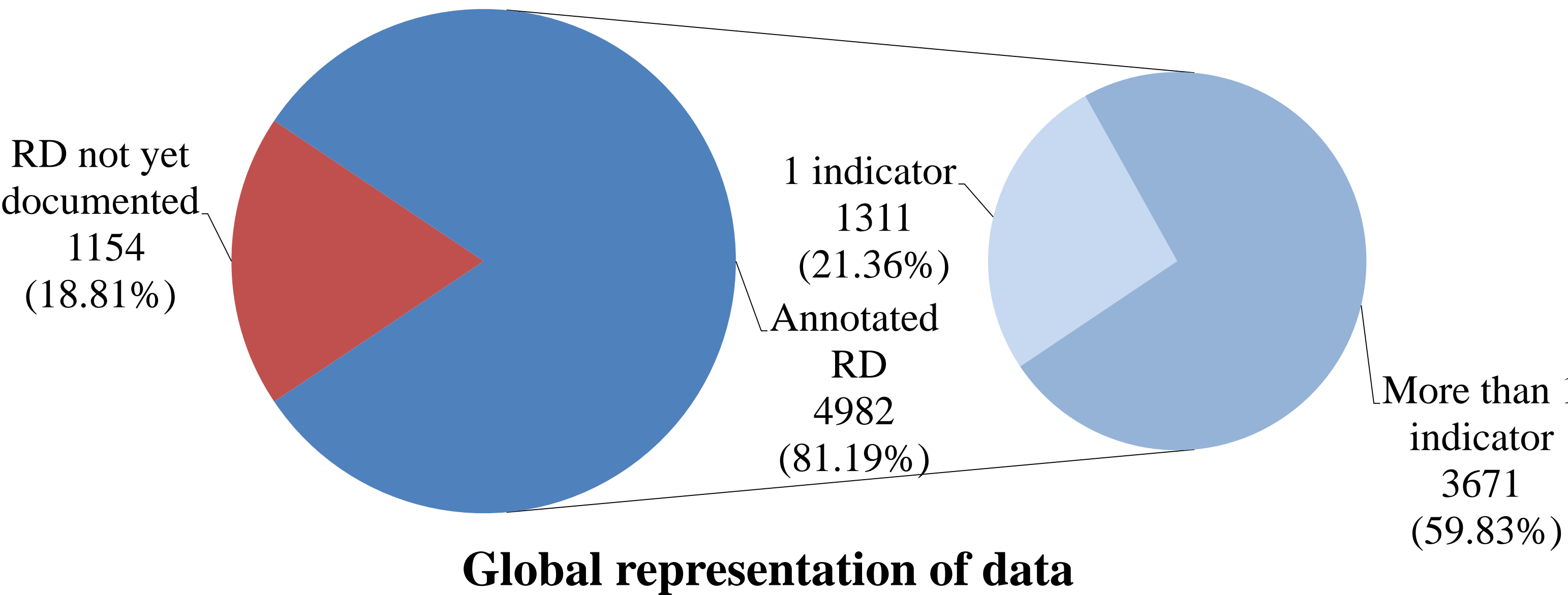


Epidemiological data life cycle

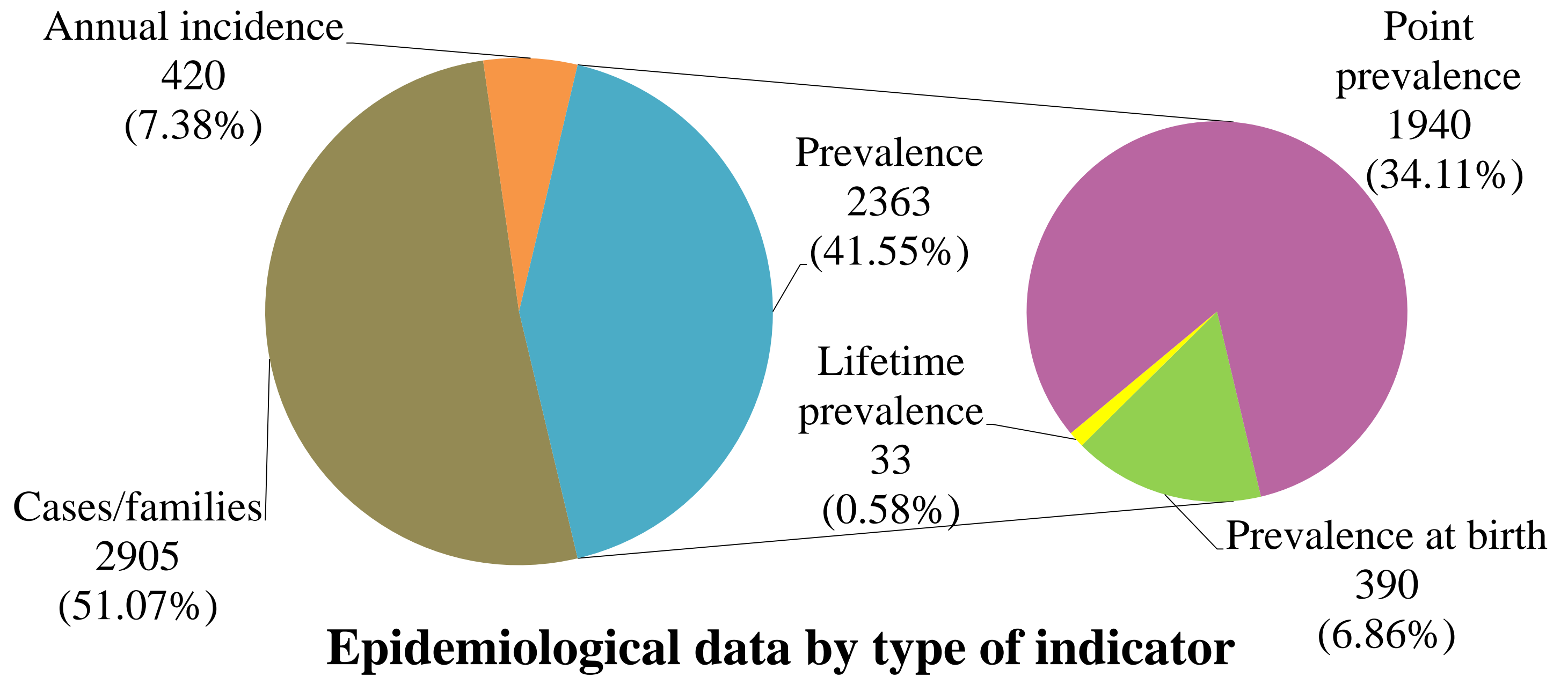


Representation of Epidemiological data in the Orphanet database

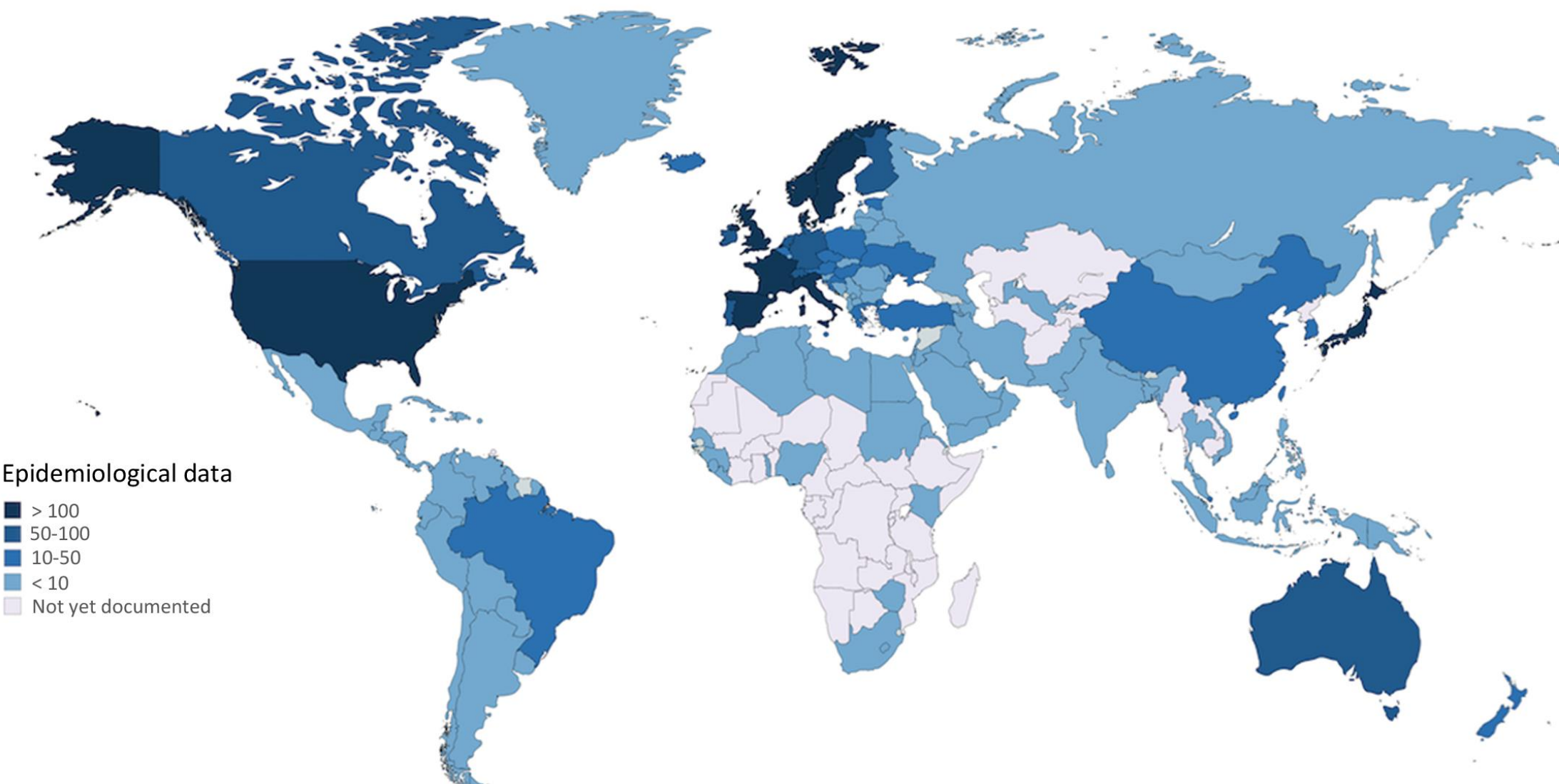
Results



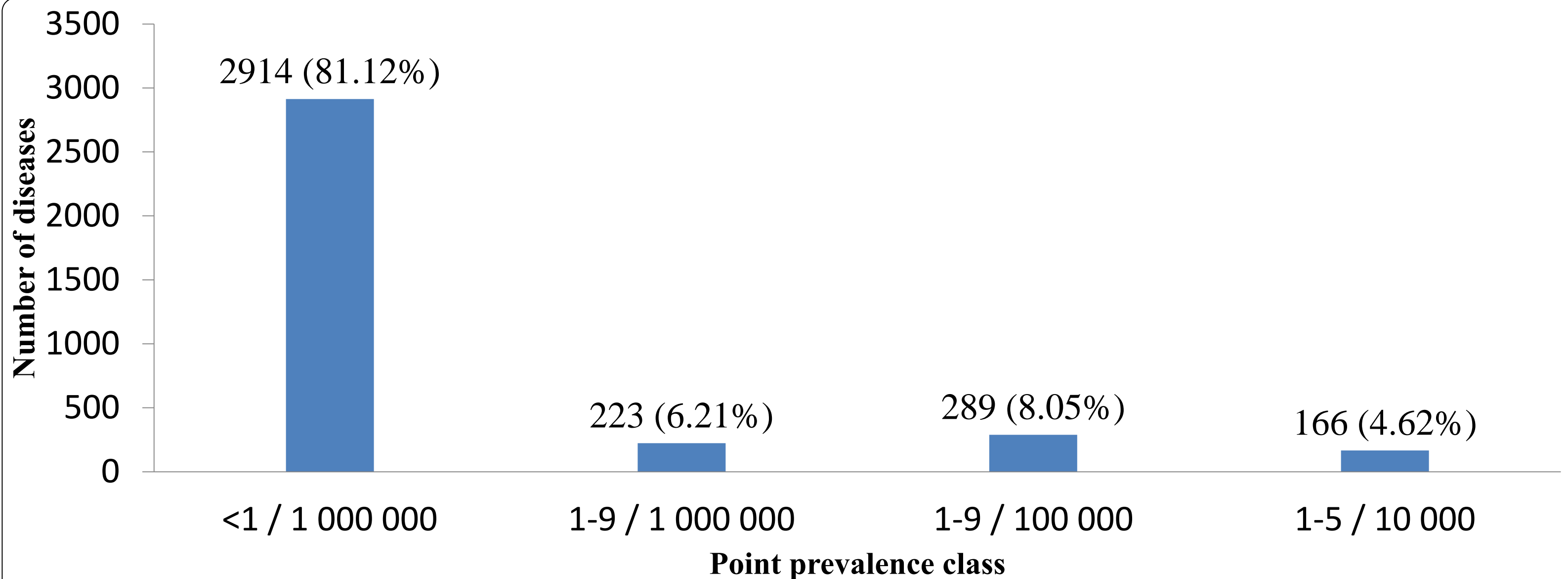
Global representation of data



Epidemiological data by type of indicator



Geographic coverage of epidemiological data



Point prevalence class distribution


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.

<h1>Polycythemia vera</h1>	
<h2>Definition</h2> <p>Polycythemia vera (PV) is an acquired myeloproliferative disorder characterized by an elevated absolute red blood cell mass caused by uncontrolled red blood cell production, frequently associated with uncontrolled white blood cell and platelet production.</p>	
<h2>ORPHA:729</h2>	
<p><i>Synonym(s):</i></p> <p>Acquired primary erythrocytosis</p> <p>Ossler-Vaquez disease</p> <p>PV</p> <p>Polycythemia rubra vera</p> <p>Vaquez disease</p>	<p><i>Prevalence:</i> 1.5/10 000</p> <p><i>Inheritance:</i> Not applicable</p> <p><i>Age of onset:</i> All ages</p> <p><i>ICD-10:</i> D45</p> <p><i>OMIM:</i> 263300</p> <p><i>UMLS:</i> C0032463</p> <p><i>MeSH:</i> D011087</p> <p><i>GARD:</i> 7472</p> <p><i>MedDRA:</i> 10036057</p>
<h2>Summary</h2>	
<h3>Epidemiology</h3> <p>Annual incidence is estimated at approximately 1/36,000-1/100,000 and prevalence at 1/3,300. PV occurs at all ages but is most common in those aged 50-70 years.</p>	

Information on a specific disorder

www.orpha.net



Orphanet Report Series
Rare Disorders collection






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Orphanet Re

Prevalence and incidence of rare diseases:
Bibliographic data

Prevalence, incidence or number of published cases
listed by diseases (in alphabetical order)

www.orpha.net www.orphadata.org

European Union
 Co-funded by
 the Health Programme
 of the European Union

Data compilation
Worldwide and Europe
(Published biannually)

[illegible]

Computational use subset of disorders/ data under DTA/service contract

www.orphadata.org

(Updated monthly)



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