Orphanet 20 years later
Lessons on Information needs and Value of Data

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Consultancy for…

- Pfizer
- Shire
- Biogen

KDIGO

Orphanet was established in 1997 in France to address identified problems

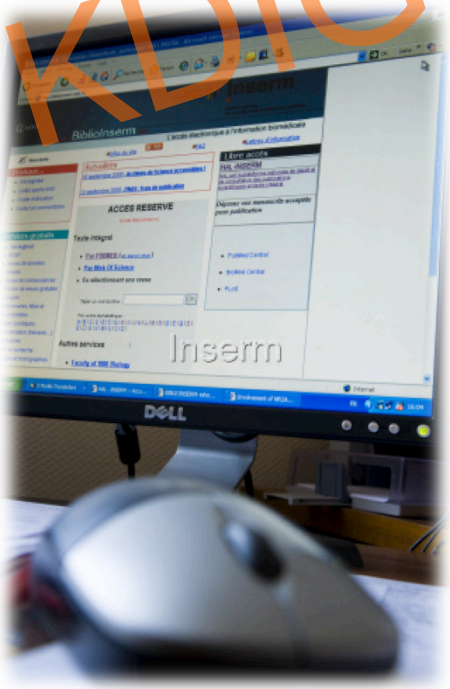
Report to the Minister of Health

Lack of information	→	Inventory and Encyclopaedia
Scarce expertise	→	Directory of experts/clinics
Too few collaborations	→	Directory of research projects
Difficult recruitment	→	Directory of clinical trials
Isolation of patients	→	Directory of patient organisations

Orphanet had Ancestors

- 1974 GENDIAG:
  - A computerized system to support the diagnosis of patients attending a genetic clinic
- 1992 GENINFO:
  - A directory of expert clinics and research groups of relevance to advance Gene Mapping

It is now the reference portal worldwide because it provides answers



orphanet

The portal for rare diseases and orphan drugs

Inserm

*Rare diseases are **rare, but** rare disease patients are **numerous***

Access our Services

Search a disease OK

- Inventory, classification and encyclopeadia of rare diseases, with genes involved
- Assistance-to-diagnosis tool
- Emergency guidelines
- Inventory of orphan drugs
- Directory of medical laboratories providing diagnostic tests
- Directory of expert centres
- Directory of ongoing research projects, clinical trials, registries and biobanks
- Directory of patient organisations
- Directory of professionals and institutions
- Newsletter
- Collection of thematic reports: Orphanet Reports Series

Read Orphanet reports

- Prevalence of Rare Diseases
- Disease registries in Europe
- European research projects & clinical networks
- Lists of Orphan Drugs
- Orphanet Activity Reports
- Satisfaction Surveys

Contribute to Orphanet

- Register your activity
- Sponsor Orphanet [↗]

Download Orphanet data

- Orphadata [↗]

Newsletter

- Read the last newsletter
- Read previous issues
- Sign up to receive the newsletter

Other documents

- Council Recommendation on an action in the field of rare diseases [↗]
- State of Art of rare diseases [↗]

Other rare diseases websites

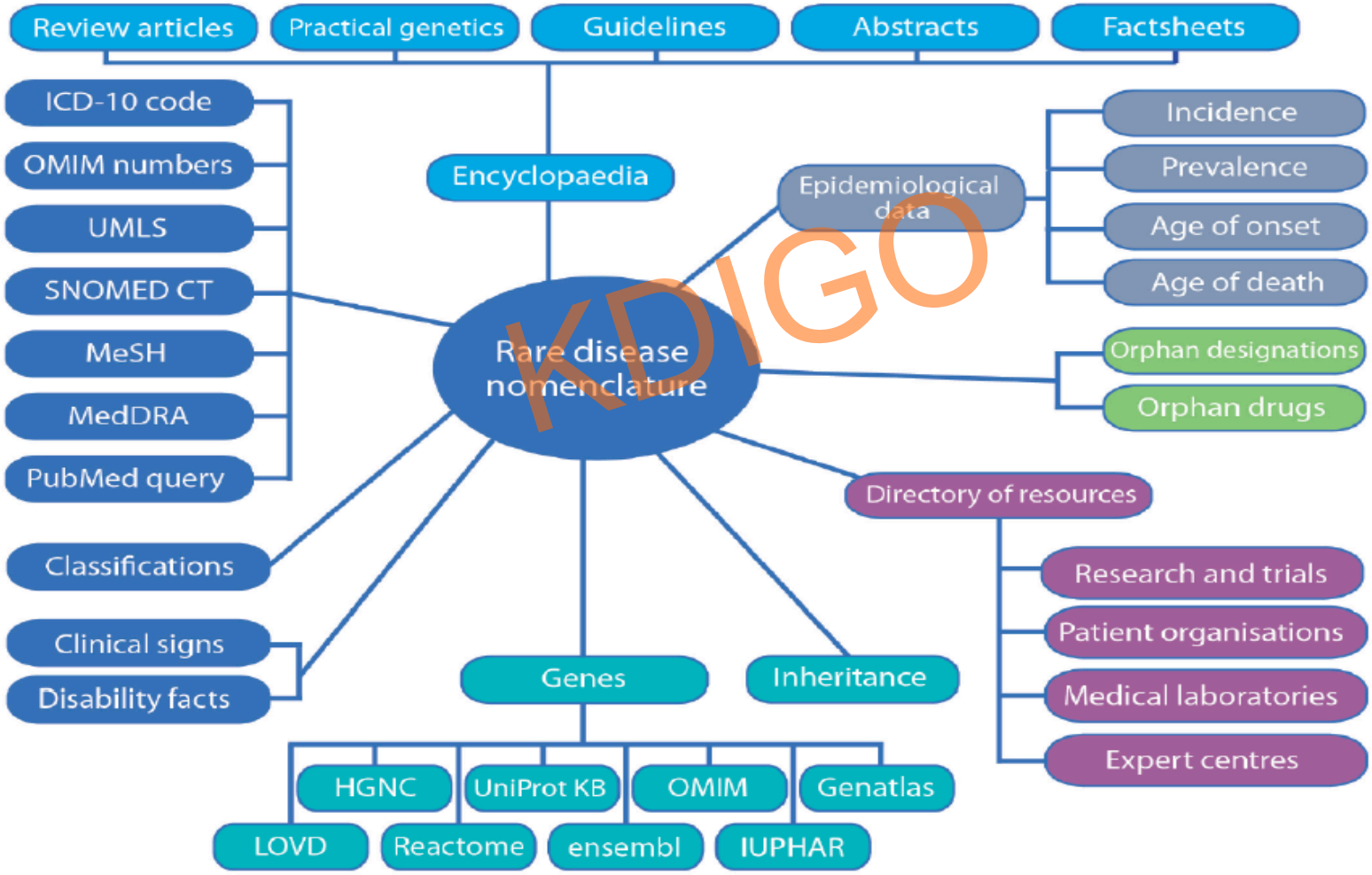
- Rare Diseases - European Commission
- EUCERD
- European Medicines Agency
- Office of rare diseases research (US)

Events

See all

OCT 8 — EpiRare International Workshop: Rare Disease and Orphan Drug Registries [↗]
8-9 Oct 2012, Roma, Italy [↗]

Content of Orphanet



Orphanet Consortium for Expert Services in 2016

The End Users

Orphanet: over 13 Million Visits from 1 million unique Users in 2015

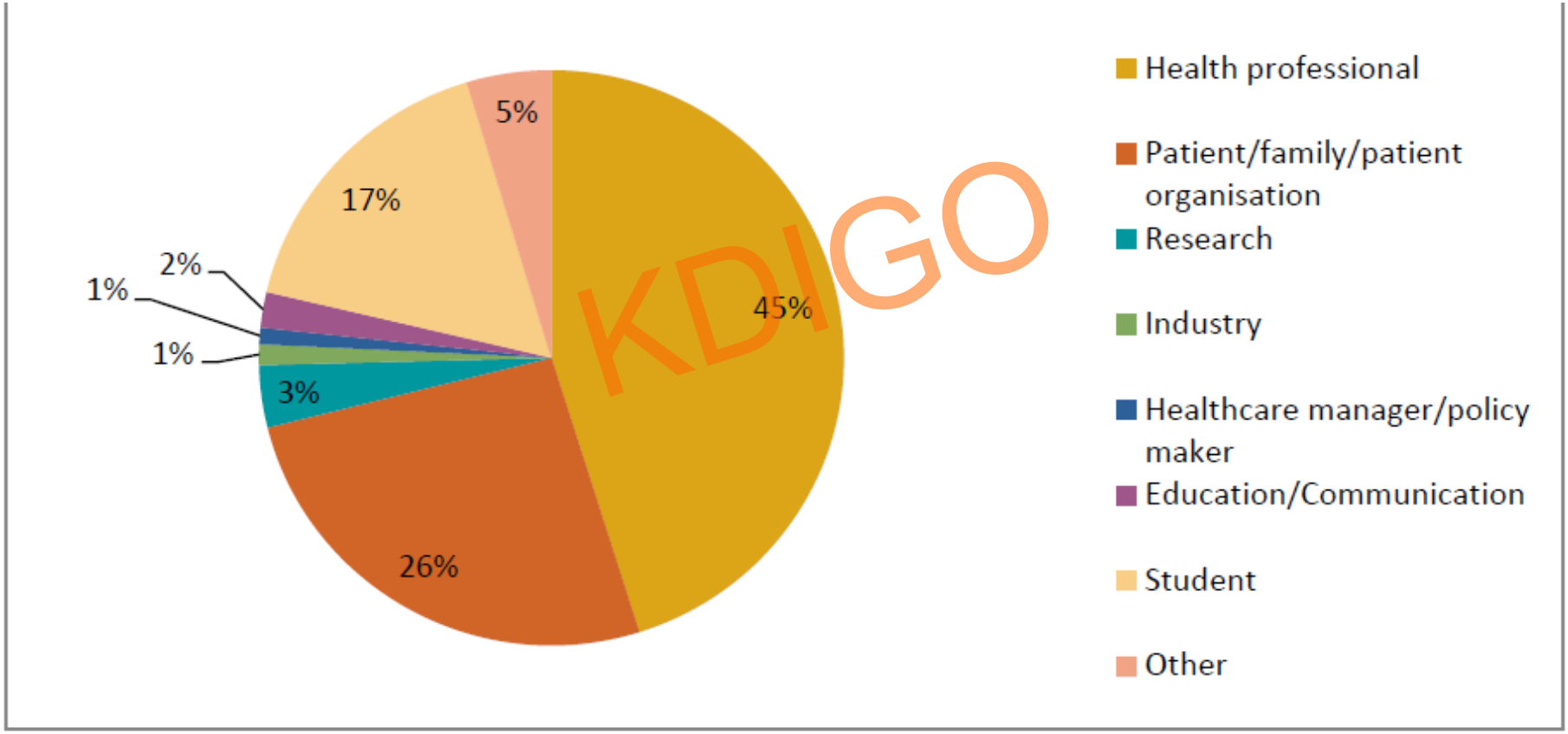
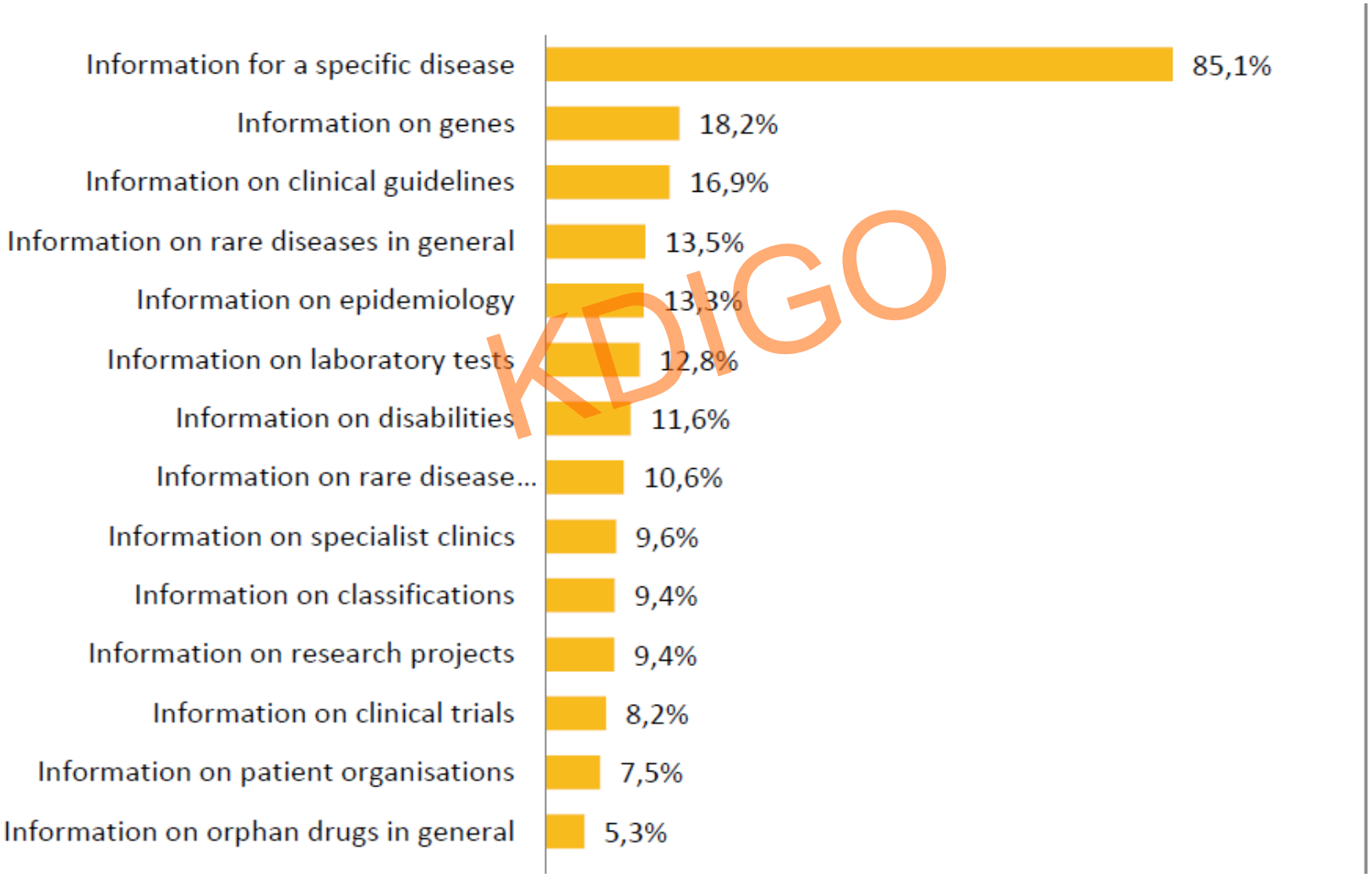


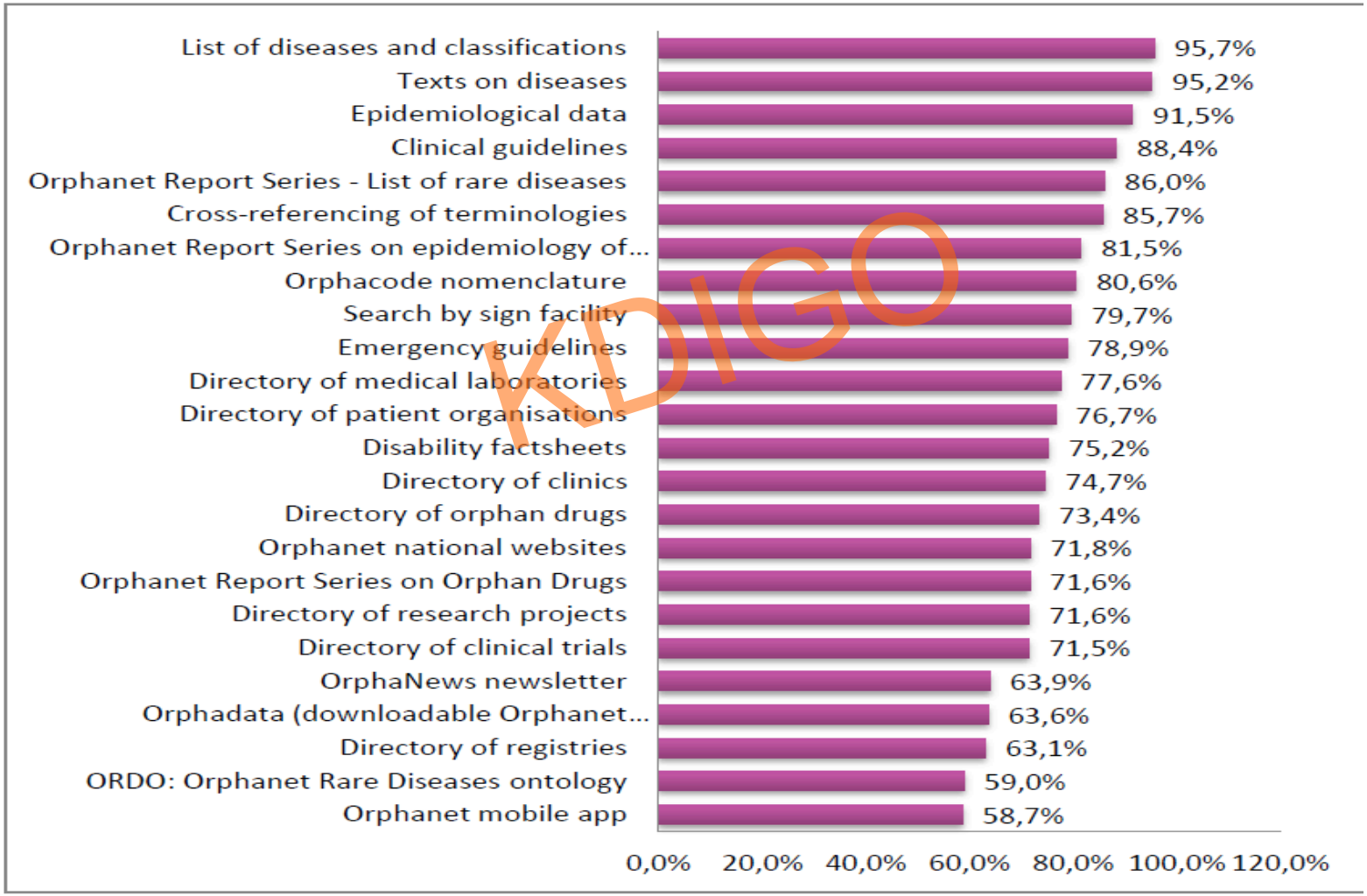
Figure 27 Types of Orphanet users (percentage of total respondents) n = 3795

Visitors from over 200 countries

Main Reason for Visiting



Utility of Orphanet services as ranked by users



20 years of Feed-back from End-Users

1- Inventory of Rare Diseases

2- Encyclopaedia

3- Inventory of Drugs (R&D and Marketed drugs)

4- Inventory of Expert services

5 - Data freely accessible to researchers for re-use

6- Communication Channels

1- Inventory of Rare Diseases

Orphanet Inventory of Diseases

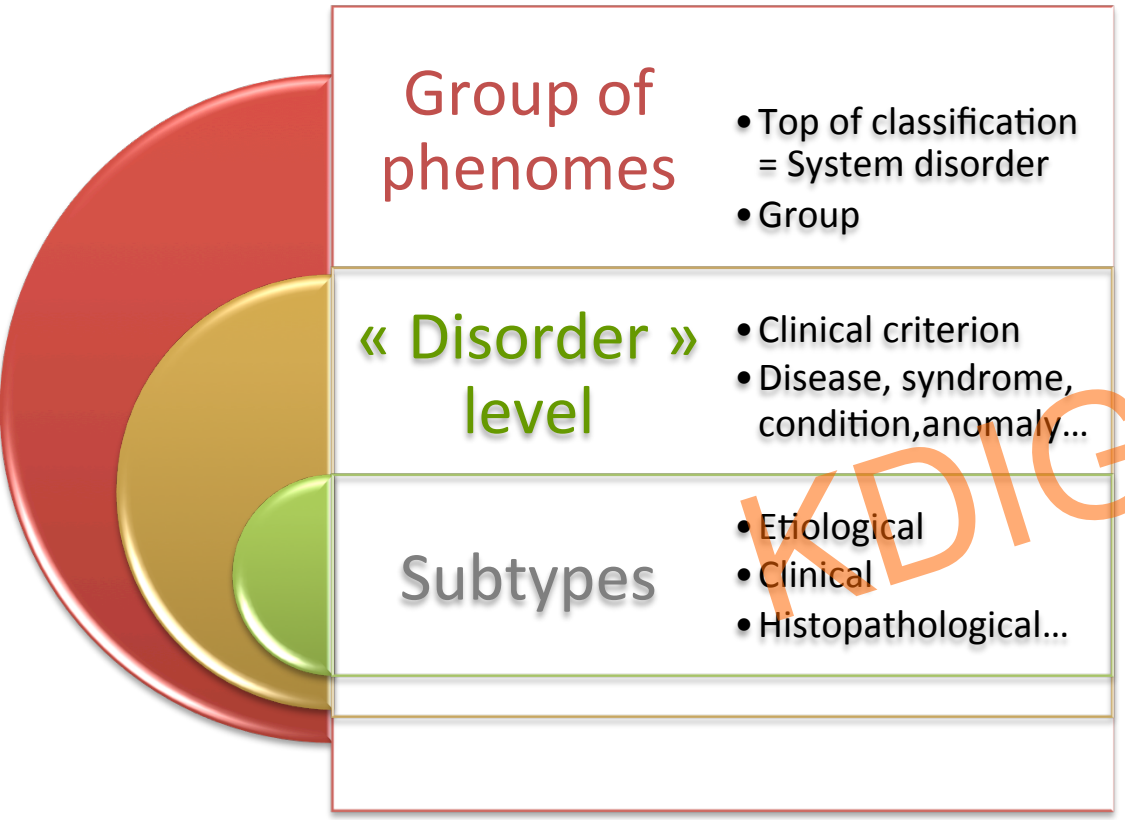
- Identity card + genes
- Unique Orpha number
    - Stable whatever the evolution of knowledge
    - Every disease is positionned in classifications

- **Suitable to code clinical activity / lab activity in information systems**

- Suitable to cluster diseases for research purpose

- Serve as a base for the revision of the International Classification of Rare Diseases at WHO

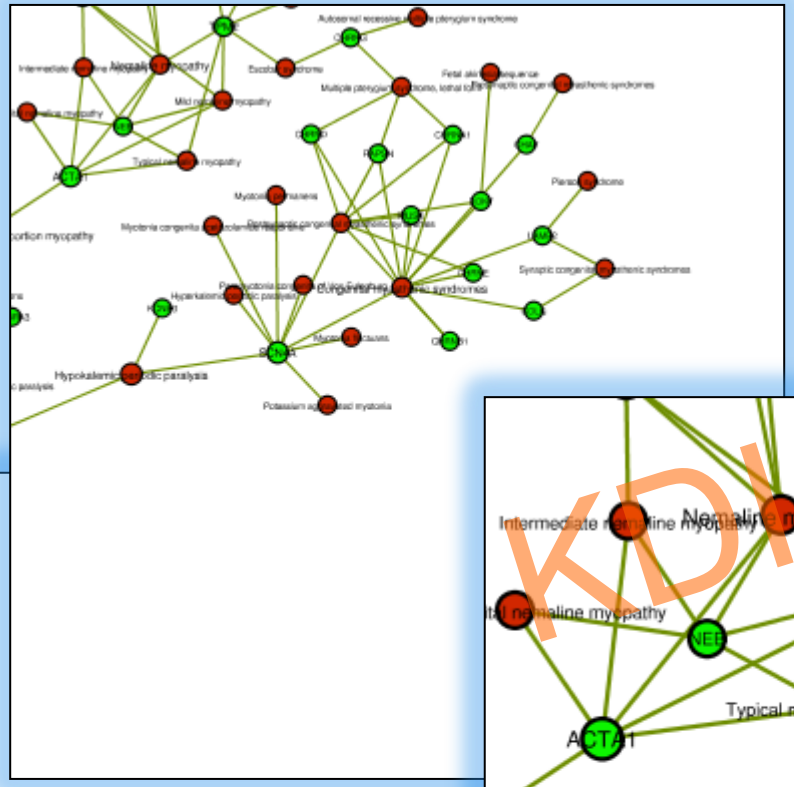
Phenomes: a continuum + zooms

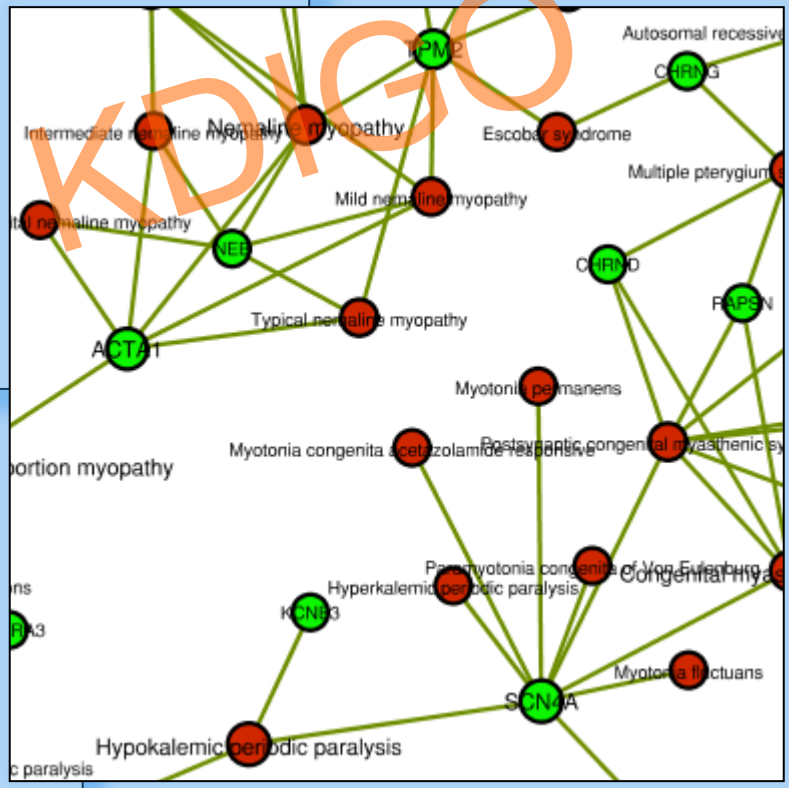


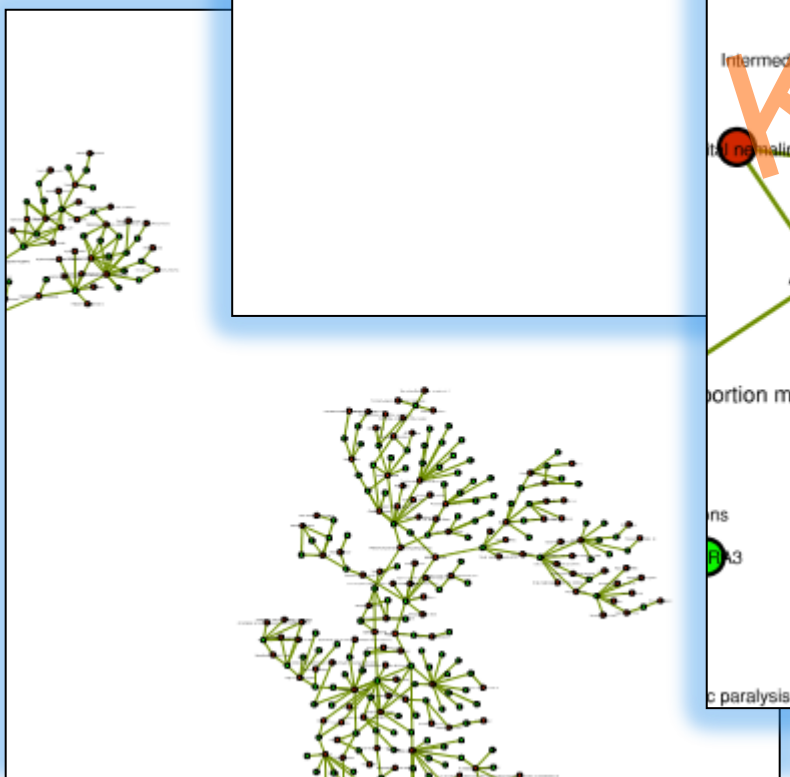
KDIGO

- Disease
- Malformation syndrome
- Morphological anomaly
- Biological anomaly
- Clinical syndrome
- Particular clinical situation

Orphan Diseasome







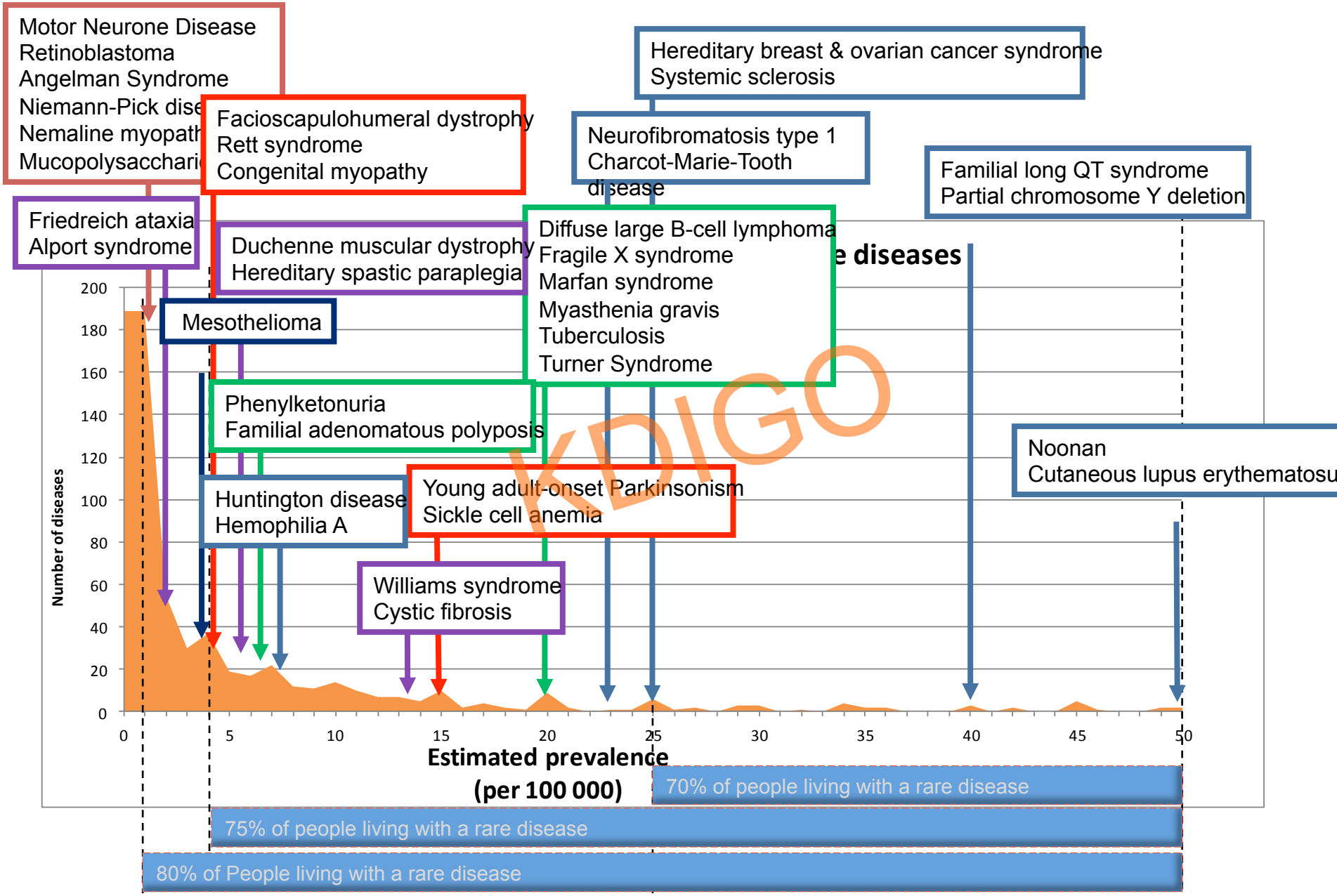
An Orphan Diseasome permits investigators to explore the orphan disease (OD) or rare disease relationships based on shared genes and shared enriched features (e.g., Gene Ontology Biological Process, Cellular Component, Pathways, Mammalian Phenotype).

The red nodes represent the orphan diseases and the green ones the related genes. A disease is connected to a gene if and only if a mutation which is responsible of the disease has been identified on this gene.

http://research.cchmc.org/od/01/index.html

Ontologies in the field of RD

- HPO: Human Phenotype Ontology
  - To describe the signs and symptoms
  - Developed at La Charité, Berlin
- ORDO: Orphanet Rare Diseases Ontology
  - To describe the entity in an aggregated manner
  - Developed at INSERM, Paris
  - Collaboration with the EBI


Motor Neurone Disease
Retinoblastoma
Angelman Syndrome
Niemann-Pick dise
Nemaline myopath
Mucopolysacchari
Facioscapulohumeral dystrophy
Rett syndrome
Congenital myopathy
Hereditary breast & ovarian cancer syndrome
Systemic sclerosis
Neurofibromatosis type 1
Charcot-Marie-Tooth
disease
Familial long QT syndrome
Partial chromosome Y deletion
Friedreich ataxia
Alport syndrome
Duchenne muscular dystrophy
Hereditary spastic paraplegia
Diffuse large B-cell lymphoma
Fragile X syndrome
Marfan syndrome
Myasthenia gravis
Tuberculosis
Turner Syndrome
e diseases
Mesothelioma
Phenylketonuria
Familial adenomatous polyposis
Noonan
Cutaneous lupus erythematosu
Huntington disease
Hemophilia A
Young adult-onset Parkinsonism
Sickle cell anemia
Williams syndrome
Cystic fibrosis
KDIGO
Number of diseases
0
20
40
60
80
100
120
140
160
180
200
0
5
10
15
20
25
30
35
40
45
50
Estimated prevalence
(per 100 000)
70% of people living with a rare disease
75% of people living with a rare disease
80% of People living with a rare disease

Contribution of each Medical Field in % of the RD total prevalence

- Dysmorphology 16.4
- Oncology 10.9
- Neurology 10.8
- Metabolism 7.4
- Haematology 6.8
- Cardiology 6.7
- Bone diseases 5.6
- Dermatology 5.4
- Rhumatology 4.5
- Endocrinology 4.5
- Ophthalmology 3.7
- Pneumology 3.5
- Infectious diseases 2.6
- Neuromuscular 2.4
- Vascular diseases 2.2
- NET * 1.8
- **Nephrology** **1.7**
- Gastroenterology 1.7
- Hepatology 1.3
- Immunology 0.3

Orphanet data 2013

* Neuroendocrine gastrointestinal tumors

5- Free Acess to Orphanet Data Since 2011

Uptake of Orphadata Files 2011 - 2015

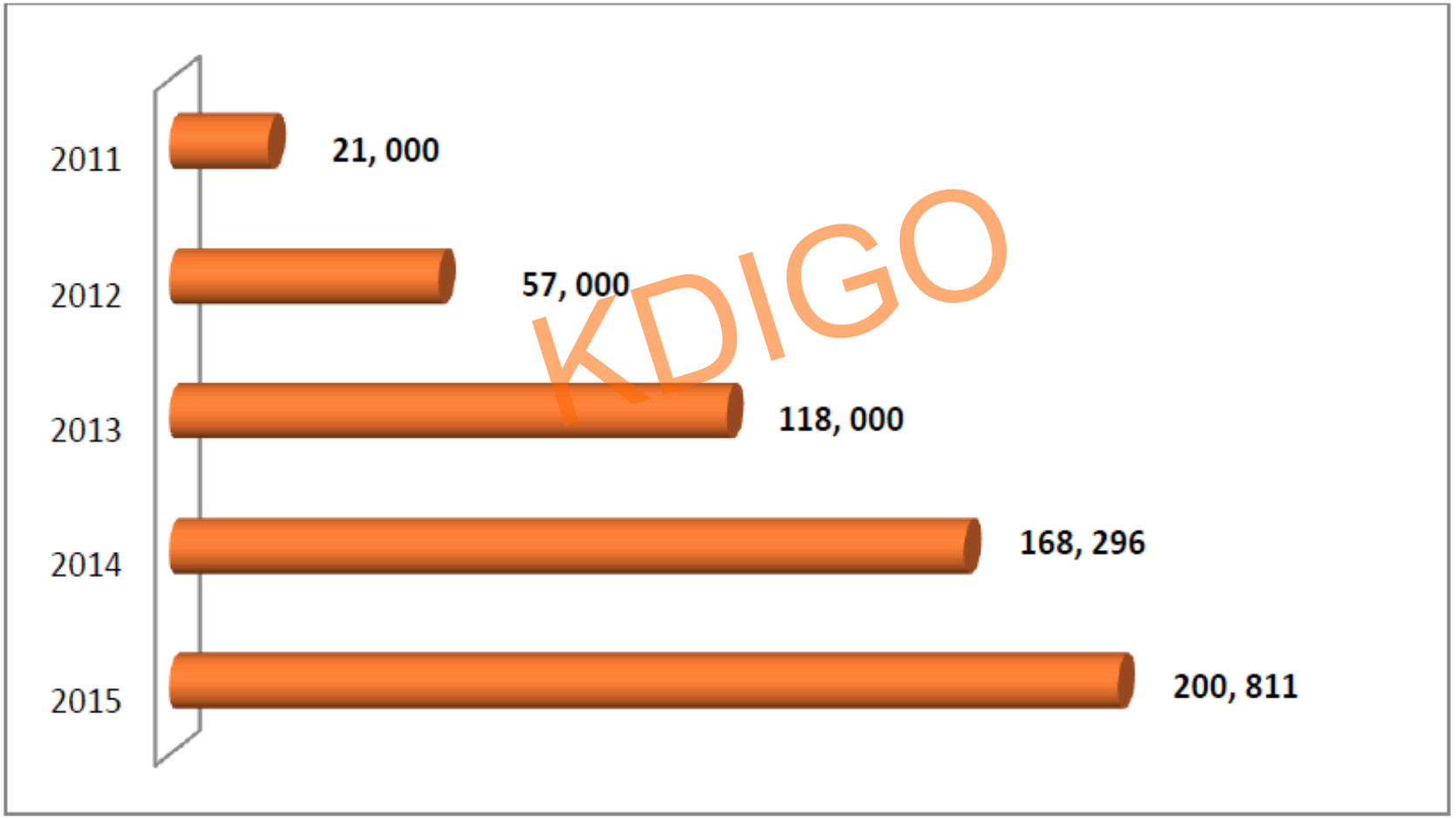


Figure 21 Number of downloads from the Orphadata website since mid 2011

Most popular products for re-use

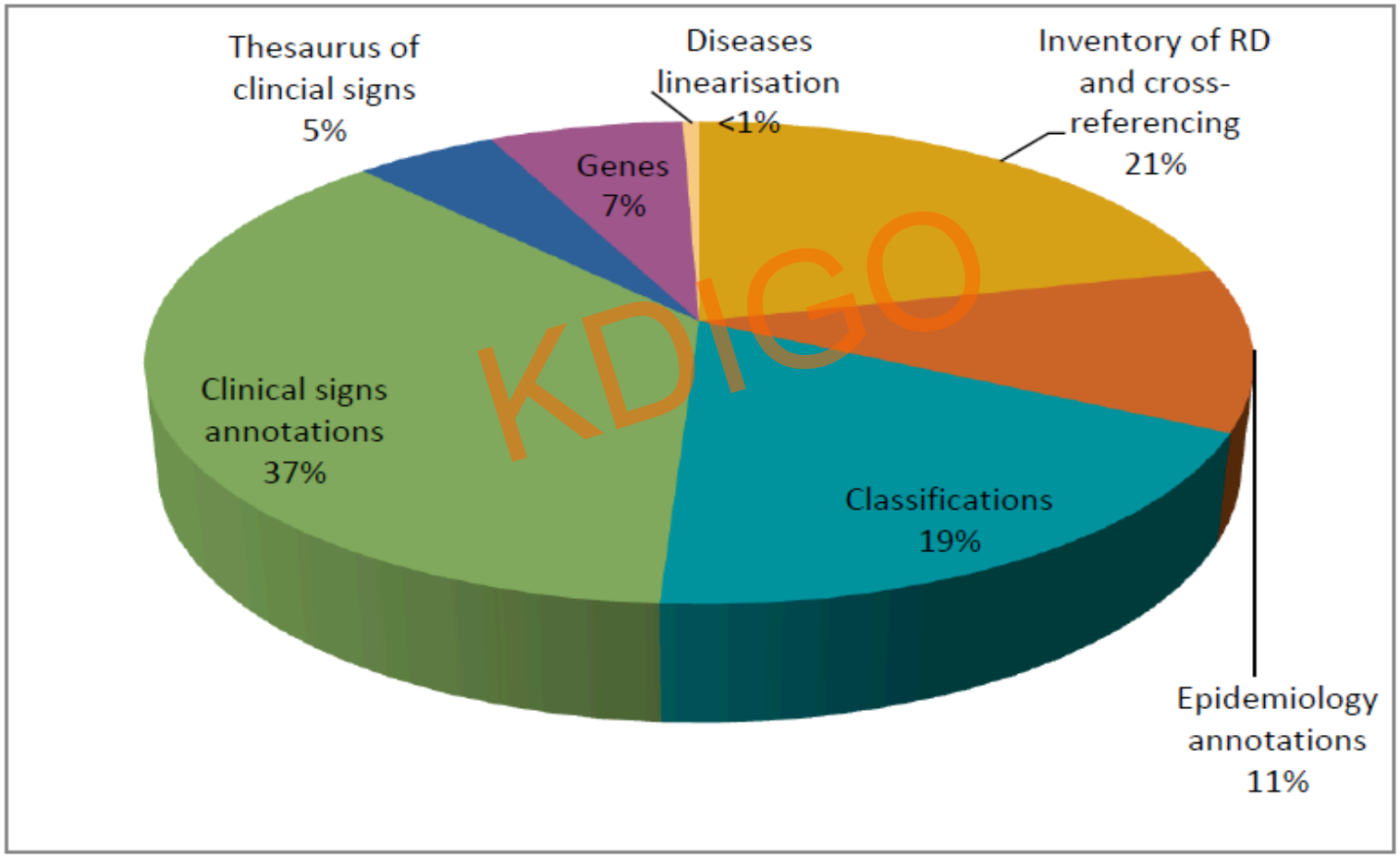


Figure 22 Distribution of the downloads of Orphadata freely available datasets in 2015

[total of 200,383 downloads]

6- Communication Channels

Orphanet Report Series

OrphaNews

Orphanet Report Series

Rare Diseases collection

January 2011

Disease Registries in Europe

Orphanet Report Series

Rare Diseases collection

November 2011 | Number 1

Prevalence of rare diseases: Bibliographic data

Listed in alphabetical order of diseases

Inserm

Orphanet Report Series

Orphan Drugs collection

January 2013

Lists of medicinal products for rare diseases in Europe

www.orpha.net

Inserm

Orphanet Report S

Rare Disea

December 2012 | Num

List of rare diseases

Listed in alphabetical order

www.orpha.net

Inserm

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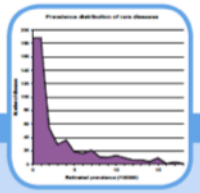
the field of rare diseases

www.orpha.net



Orphanet Report Series

Rare Diseases collection

November 2011 | Number 2

Prevalence of rare diseases: Bibliographic data

Listed in order of decreasing prevalence or number of published cases

www.orpha.net

Inserm

Orphanet Report Series Number of Downloads

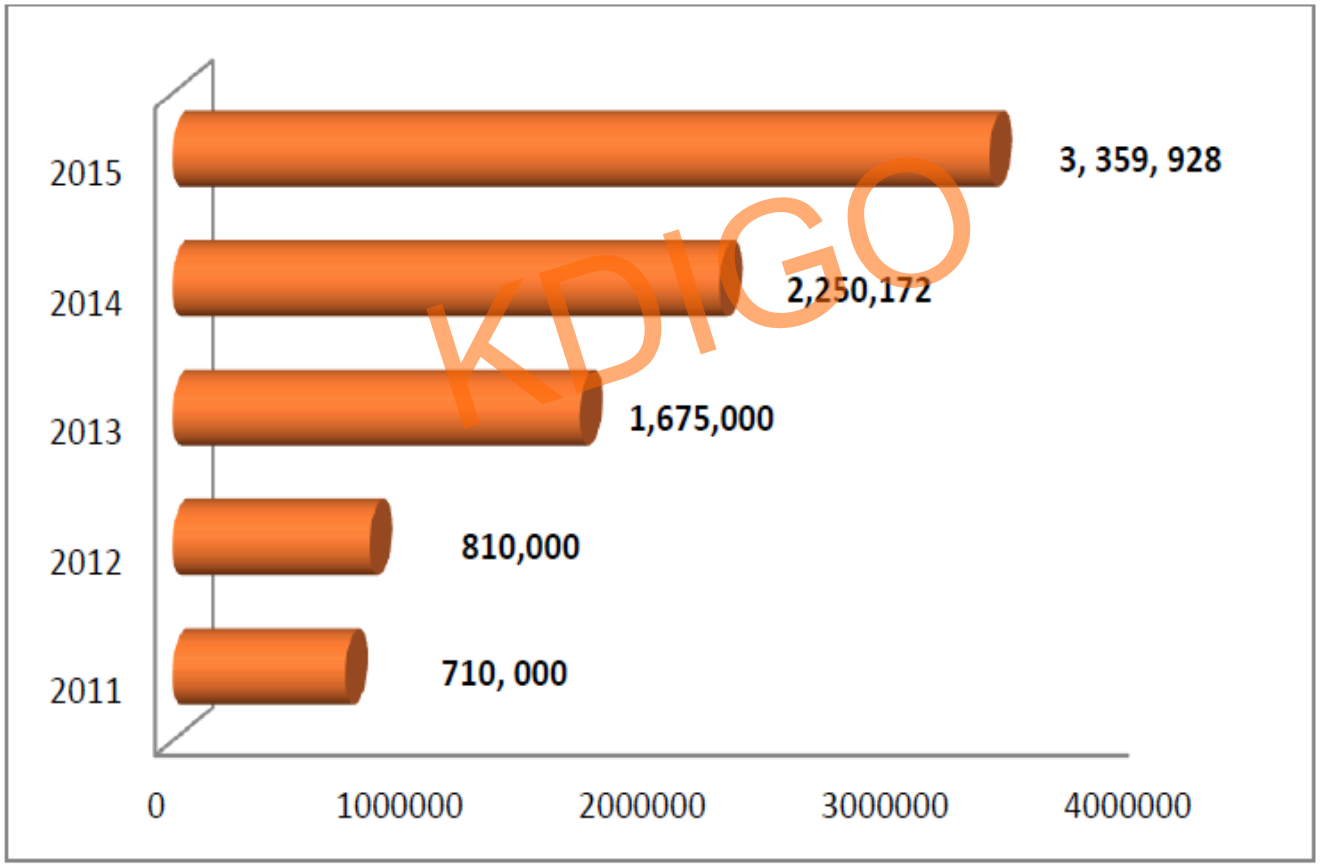


Figure 25 Number of downloads of the Orphanet Report Series in all languages since 2010

Conclusion

- Enormous appetite for authoritative information vs Social Media vs raw data
- Collaborative effort
  - Clinical guidelines
  - Annotation signs and symptoms
  - Expert clinics
  - Curation of Data
- Free access is a pre-request
  - Creative Commons

Thank you!