



# orphanet

[www.orpha.net](http://www.orpha.net)

The **reference  
source**

for documentation  
on **rare diseases**

# Orphanet was established in 1997 to address identified problems.....

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

**SIMPLE SEARCH**

→ **OK**

> Alphabetical list of rare diseases

**OTHER SEARCH OPTION(S)**

- > **Orphan drugs**
- > **Research and trials**
- > **Diagnostic tests**
- > **Patient organisations**
- > **Clinics**
- > **Directory of resources**

**ORPHANET DATA**

Diseases	: 7242
Clinics	: 3017
Laboratories	: 2838
Professionals	: 10136
Daily visitors	: 22558

- RARE DISEASES**
- > Information about a disease
  - > Alphabetical list
  - > Search by clinical sign
  - > Emergency guidelines
  - > Patient encyclopaedia
  - > Professional encyclopaedia
  - > About Rare Diseases
- RESOURCES DIRECTORY**
- > Clinics
  - > Reference centres
  - > Clinical laboratories
  - > Research projects
  - > Registries / databases
  - > Professionals
  - > Patient organisations
  - > EuroGentest
  - > Register your activity
- ORPHAN DRUGS**

**Improve** the quality of medical care for Rare Diseases.

**Provide** adapted services for the rare diseases community

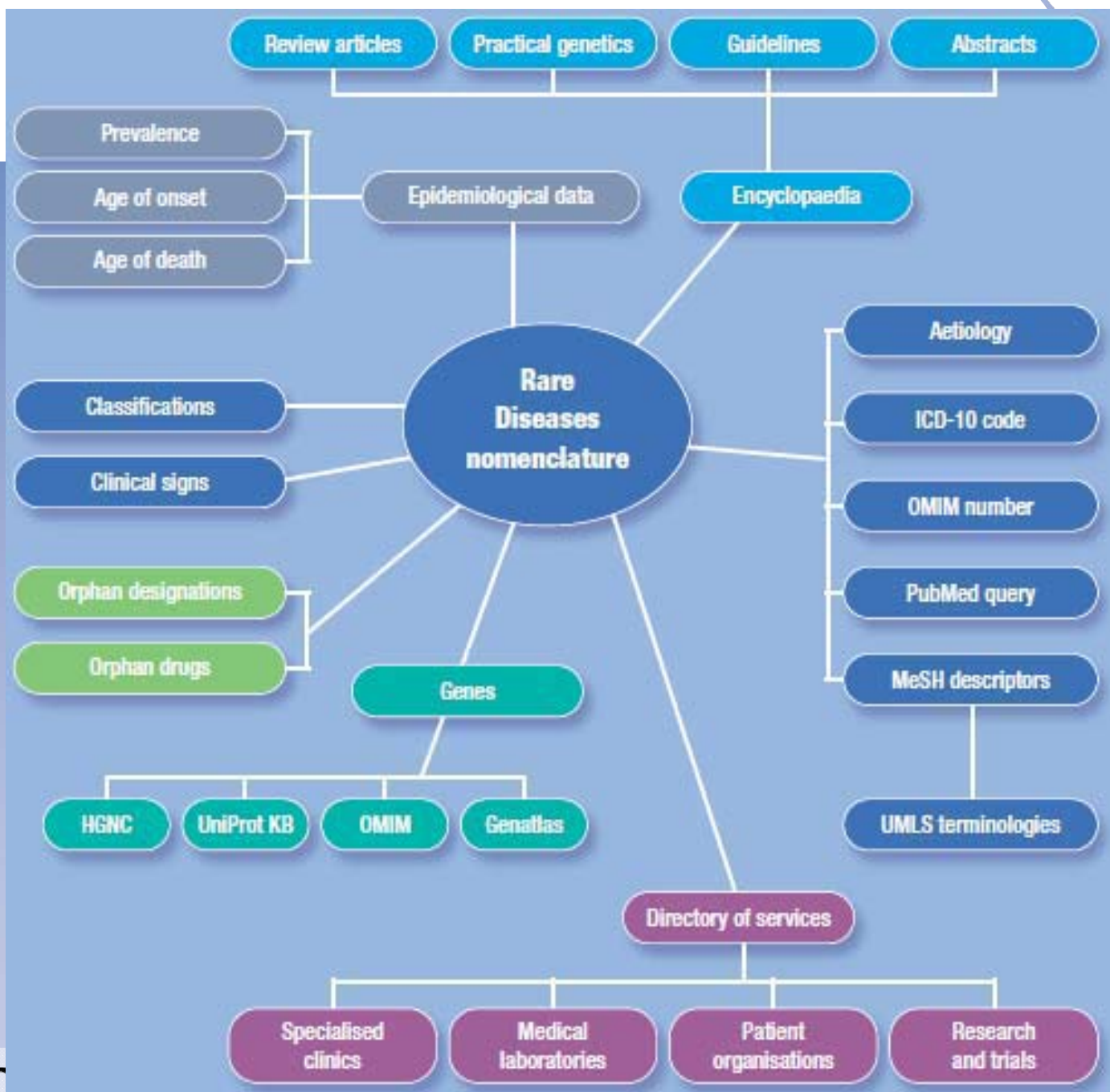
[About Orphanet](#) | [Quality charter](#)  
[Register your activity](#)



- Services for professionals**
- > Professional encyclopaedia
  - > Search by clinical sign
  - > Emergency guidelines
  - > Orphanet Journal Of Rare Diseases
  - > Newsletters
  - > Education tools
  - > Register your activity
  - > OrphanXchange

- Services for patients**
- > Patient encyclopaedia
  - > Information about a disease
  - > Patient organisations
  - > Clinics
  - > Participate in clinical research
  - > Contact other patients / families
  - > Training sessions
  - > Newsletters

- NEWS**
- .....
- [Orphanet's 10th birthday Conference presentation highlights](#)
- .....
- [EC Rare Disease webpages](#)
- .....
- [French National Plan for Rare Diseases](#)
- .....



# Indexation by clinical signs



13 Result(s)

- Angelman syndrome
- Camitine palmitoyl transferase 1 deficiency
- Cerebellar hypoplasia
- Cutis verticis gyrata - intellectual deficit
- Intellectual deficit, X-linked, South African type
- Lipodystrophy - intellectual deficit - deafness
- Menkes disease
- Mitochondrial encephalomyopathy aminocidopathy
- Neuronal ceroid lipofuscinosis
- Pelizaeus-Merzbacher disease
- Prader-Willi syndrome
- Rett syndrome
- Smith-Magenis syndrome

Results are displayed as a list of diseases with these clinical signs

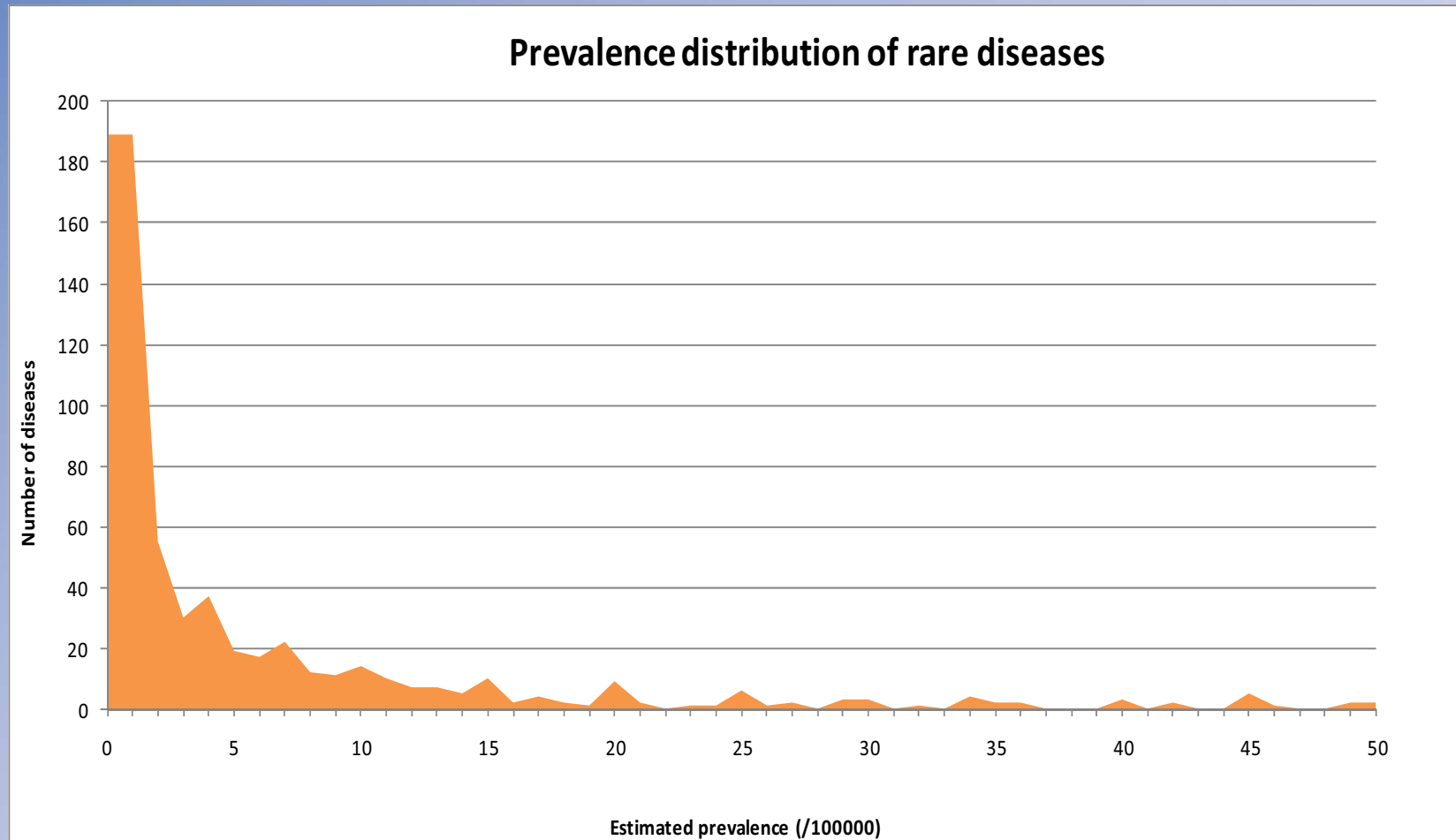
- [+] Skull/cranial anomalies
- [+] Face/facial anomalies
- [+] Anomalies of eyes and vision
- [-] Eyebrows anomalies
  - Absent/decreased/thin eyebrows
  - Thick/bushy eyebrows
  - Synophris/synophrys
  - High arched eyebrows
  - Flared eyebrows
  - Lateral thinning of eyebrows
  - Darker than expected eyebrows
- [+] Anomalies of eyelids, eyelashes and lacrimal system
- [+] Anomalies of nose and olfaction
- [+] Anomalies of mouth, lip and philtrum
- [+] Anomalies of tongue, gingiva and oral mucosa
- [+] Palate anomalies
- [+] Anomalies of teeth and dentition
- [+] Anomalies of ear and hearing

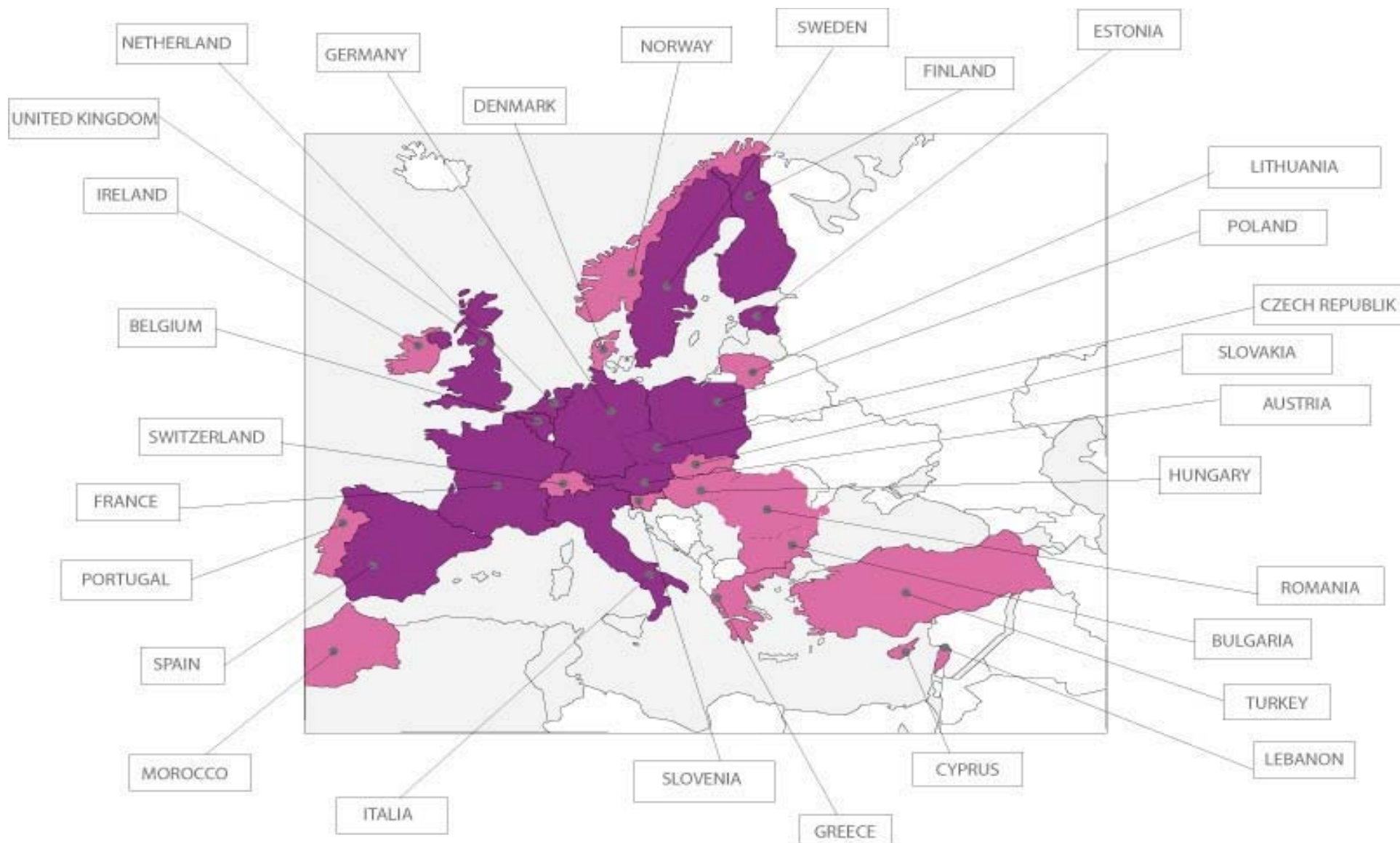
Choose from a thesaurus of 1,360 clinical signs

# Contribution of RD categories in % of total prevalence

● Dysmorphology	16.40	● Ophthalmology	3.72
● Oncology	10.87	● Pneumology	3.45
● Neurology	10.78	● Infectiology	2.59
● Metabolism	7.39	● Neuromuscular	2.42
● Haematology	6.82	● Vascular disease	2.21
● Cardiology	6.69	● Ear-Nose-Throat	1.79
● Bone	5.55	● Nephrology	1.65
● Dermatology	5.36	● Gastroenterology	1.65
● Internal Medicine	4.53	● Hepatology	1.34
● Endocrinology	4.46	● Immunology	0.32

# Distribution of prevalence rates

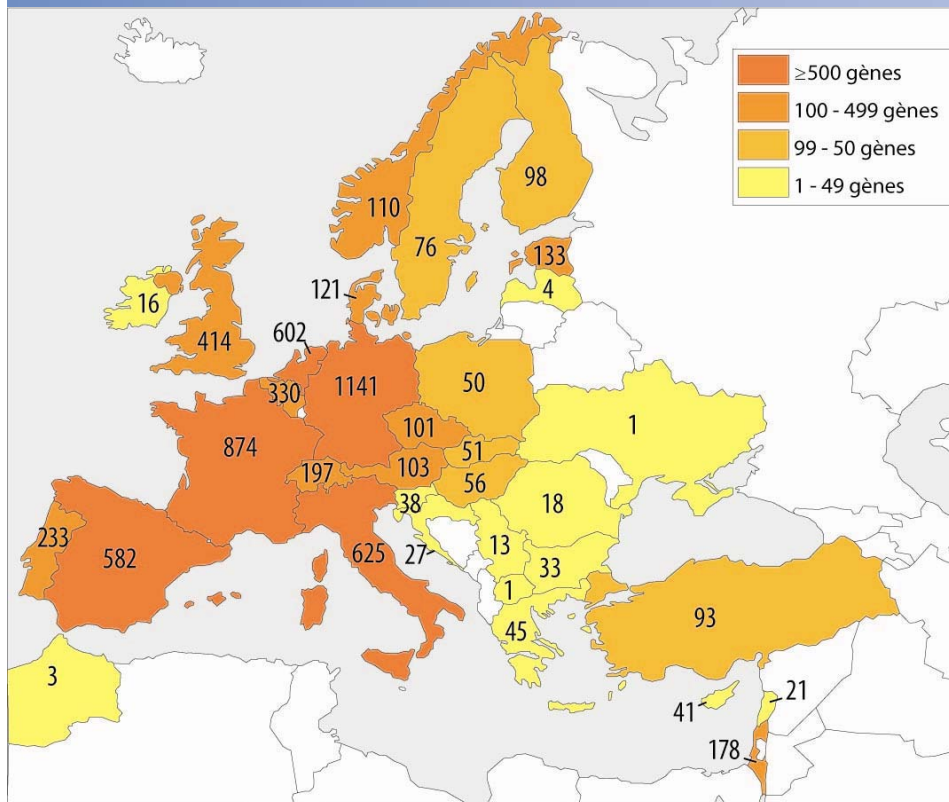




Supported by DG Sanco (Orphanet contract)  
 Supported by DG Research (RDPlatform contract)

# Diagnostic tests for genetic diseases

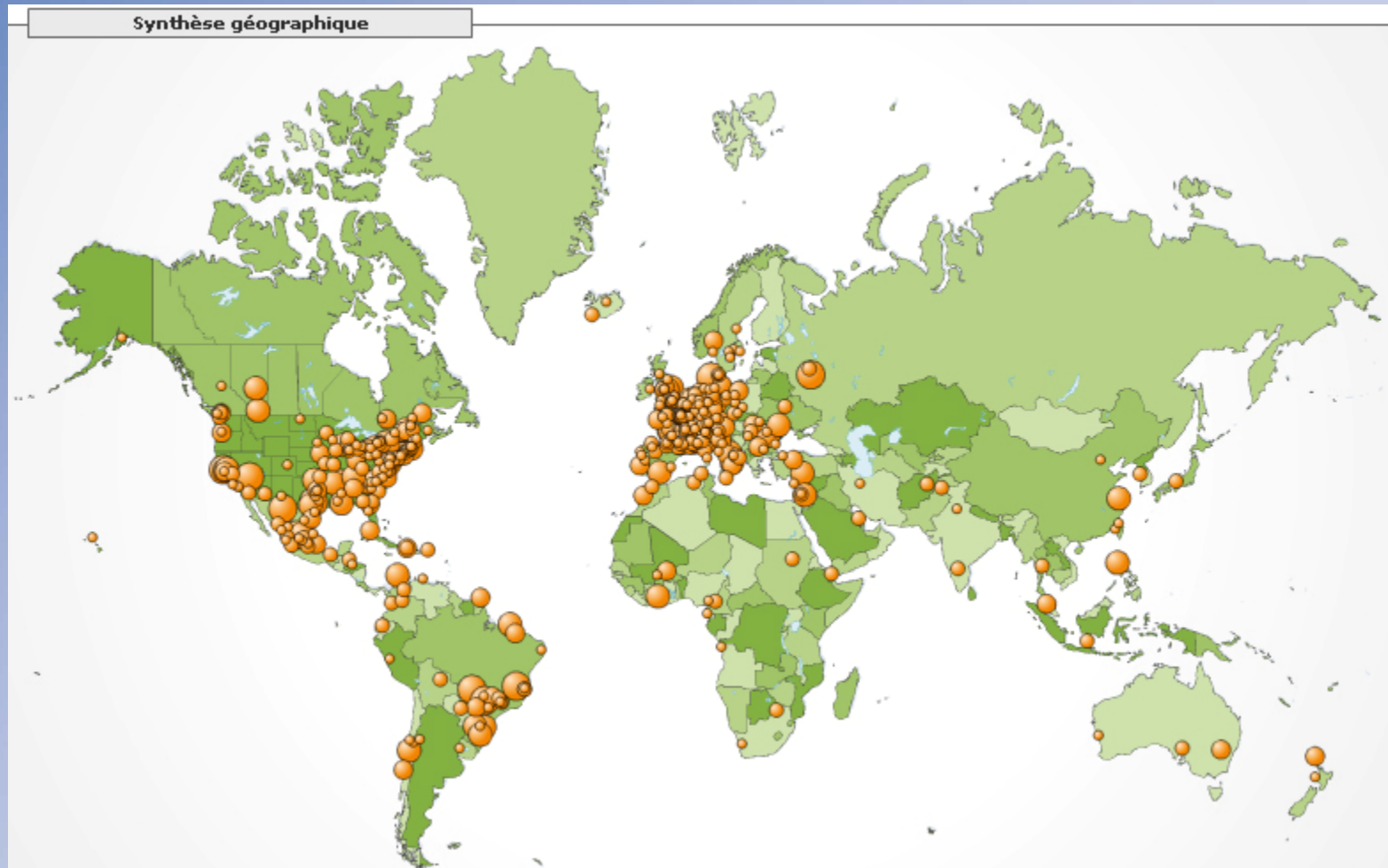
## Number of genes tested by country



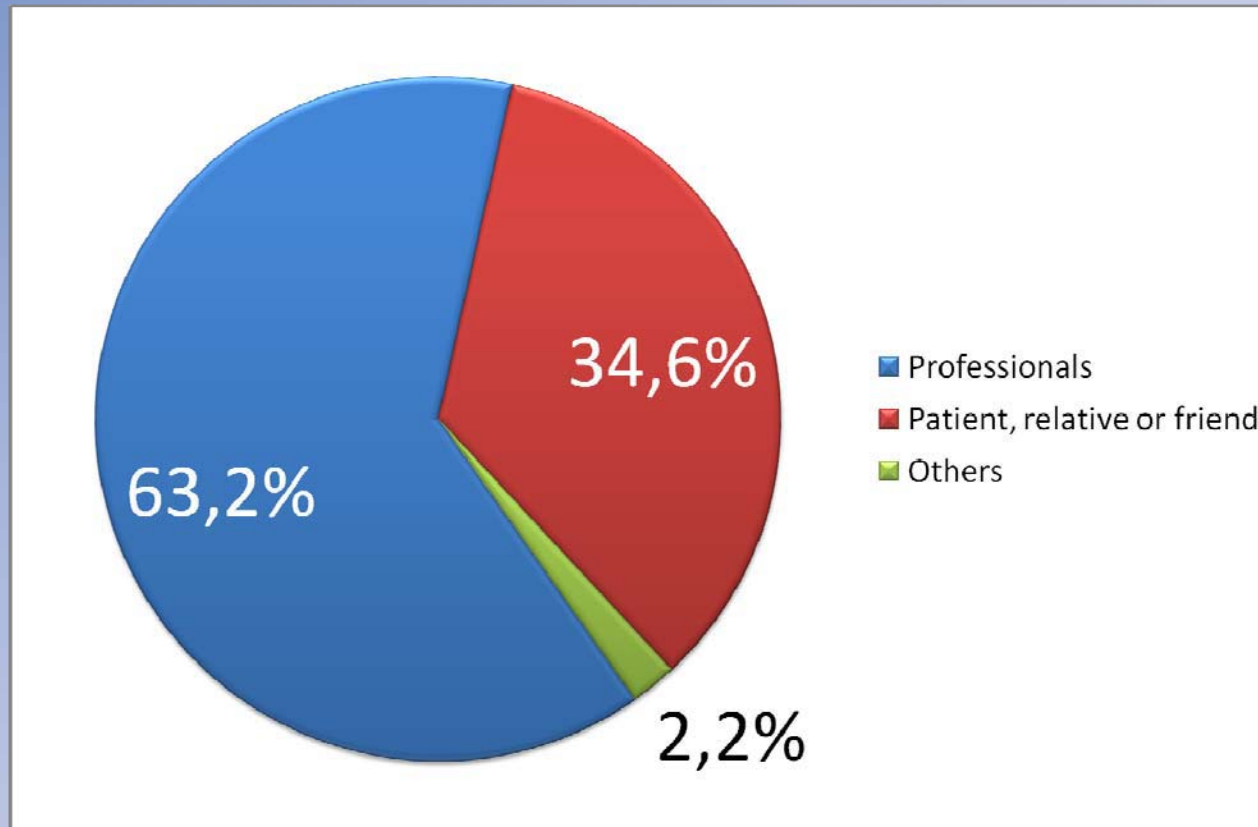
## Top 25 of diseases tested in the greatest number of European countries

Disease	Number of countries
Cystic fibrosis	32
Hemochromatosis	27
Duchenne and Becker muscular dystrophy	26
Fragile X syndrome	26
Nonsyndromic genetic deafness	25
Angelman syndrome	24
Familial breast cancer	24
Chromosome Y deletion	24
Huntington disease	24
Prader-Willi syndrome	24
Familial nonpolyposis colon cancer	23
Congenital factor II deficiency	23
Friedreich ataxia	23
Phenylketonuria	23
Homocystinuria due to methylenetetrahydrofolate reductase deficiency	22
Steinert myotonic dystrophy	22
Congenital factor V deficiency	21
Proximal spinal muscular atrophy	21
Achondroplasia	20
Beta-thalassemia	20
Chronic myeloid leukemia	20
Monosomy 22q11	20
Multiple endocrine neoplasia, type 2	20
Rett syndrome	20
Williams syndrome	20

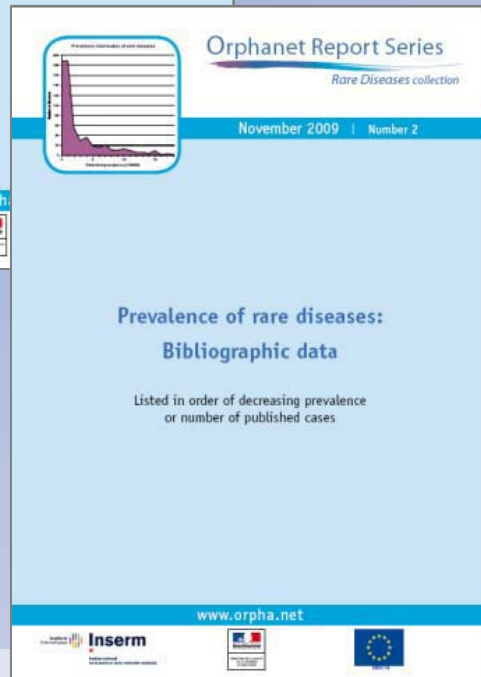
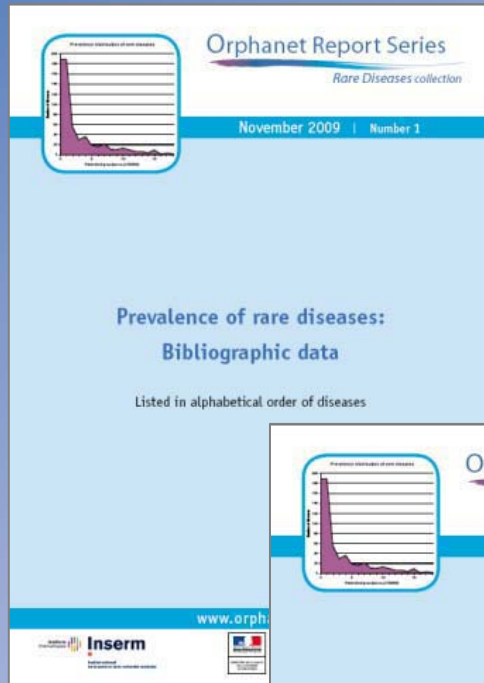
**20 000** accesses daily  
from **over 200** countries



# Orphanet Users



# Documents available on orpha.net



# OrphaNews: to stay updated on scientific and political facts: 12,000 readers



**Editorial**

**Olé! Spain officially launches national rare disease strategy and inaugurates specialised centre for social needs**

The official launch of Spain's rare disease strategy took place on 20 October. As was reported in the 10 July 2009 issue of *OrphaNews Europe*, the Spanish Ministry of Health this summer announced the establishment of a national strategy for its estimated 3 million rare disease patients and their families. The Spanish plan is essentially a coordination tool based upon seven strategic lines that will encompass the domains of information; prevention and early detection; healthcare coordination; treatment (including advanced medicinal products, orphan drugs, and related materials); social services; research; and training. The elements defined in the Spanish strategy follow the recommendations delineated by the European Council Recommendation on an Action in the Field of Rare Diseases. The Spanish strategy is available for consultation [online](#) in Spanish language.

**CENTRO DE REFERENCIA ESTATAL DE ATENCIÓN A PERSONAS CON ENFERMEDADES RARAS Y SUS FAMILIAS**

Under the Social Services element of the strategy, a national **centre** for rare disease patients has been established and was officially inaugurated by Spanish Minister of Health and Social Policy, Trinidad Jiménez on 30 September in Burgos, Spain. The residential and outpatient centre offers specialised social support services for patients and their caregivers, particularly facilitating personal autonomy and social participation for rare disease patients.