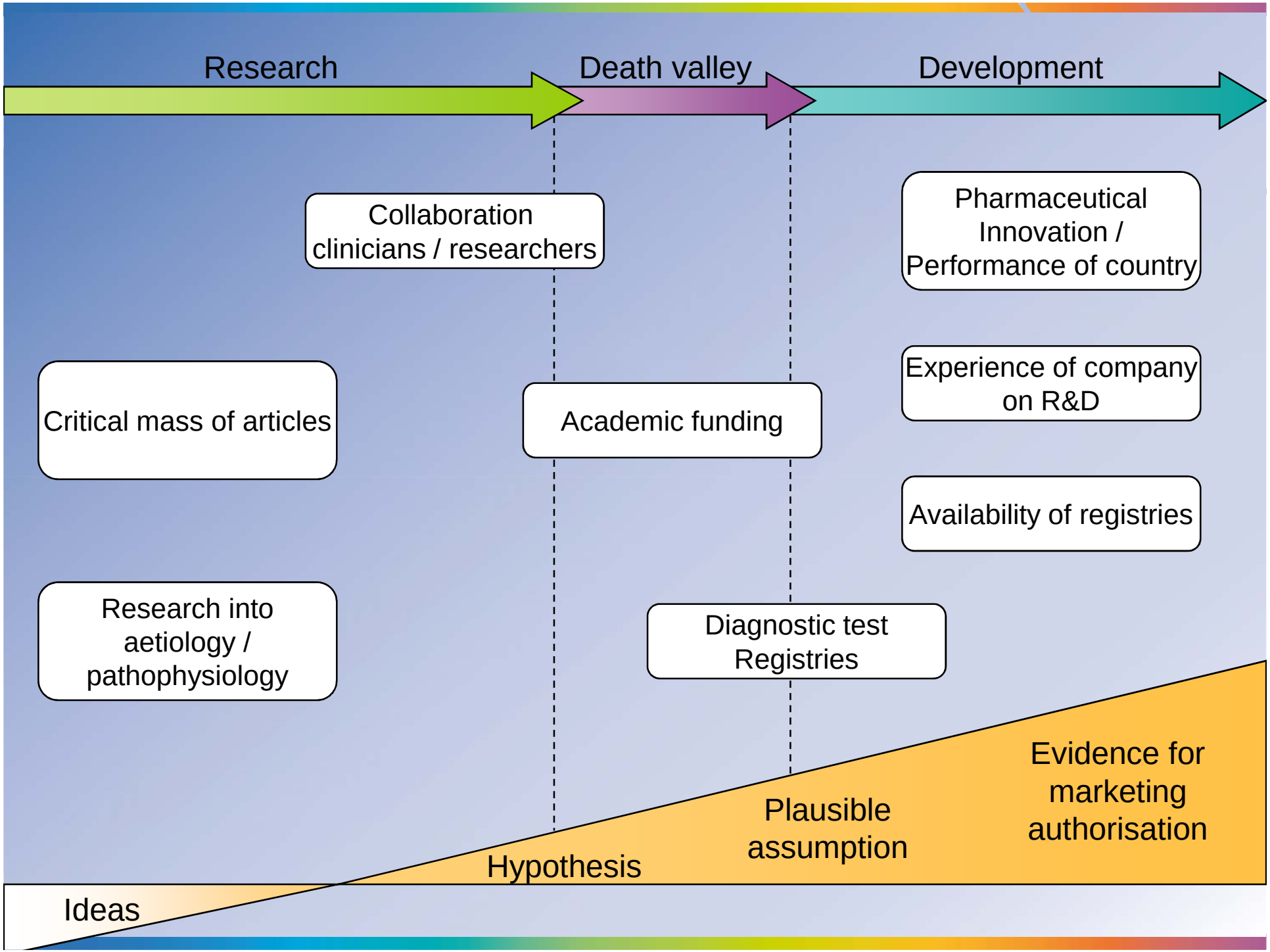
RDPlatform

Research activities in Europe: Trends and determinants

Rare Disease Day
Brussels, 1 March 2010


Research
Death valley
Development
Collaboration clinicians / researchers
Pharmaceutical Innovation / Performance of country
Critical mass of articles
Academic funding
Experience of company on R&D
Availability of registries
Research into aetiology / pathophysiology
Diagnostic test Registries
Evidence for marketing authorisation
Plausible assumption
Hypothesis
Ideas

Goals

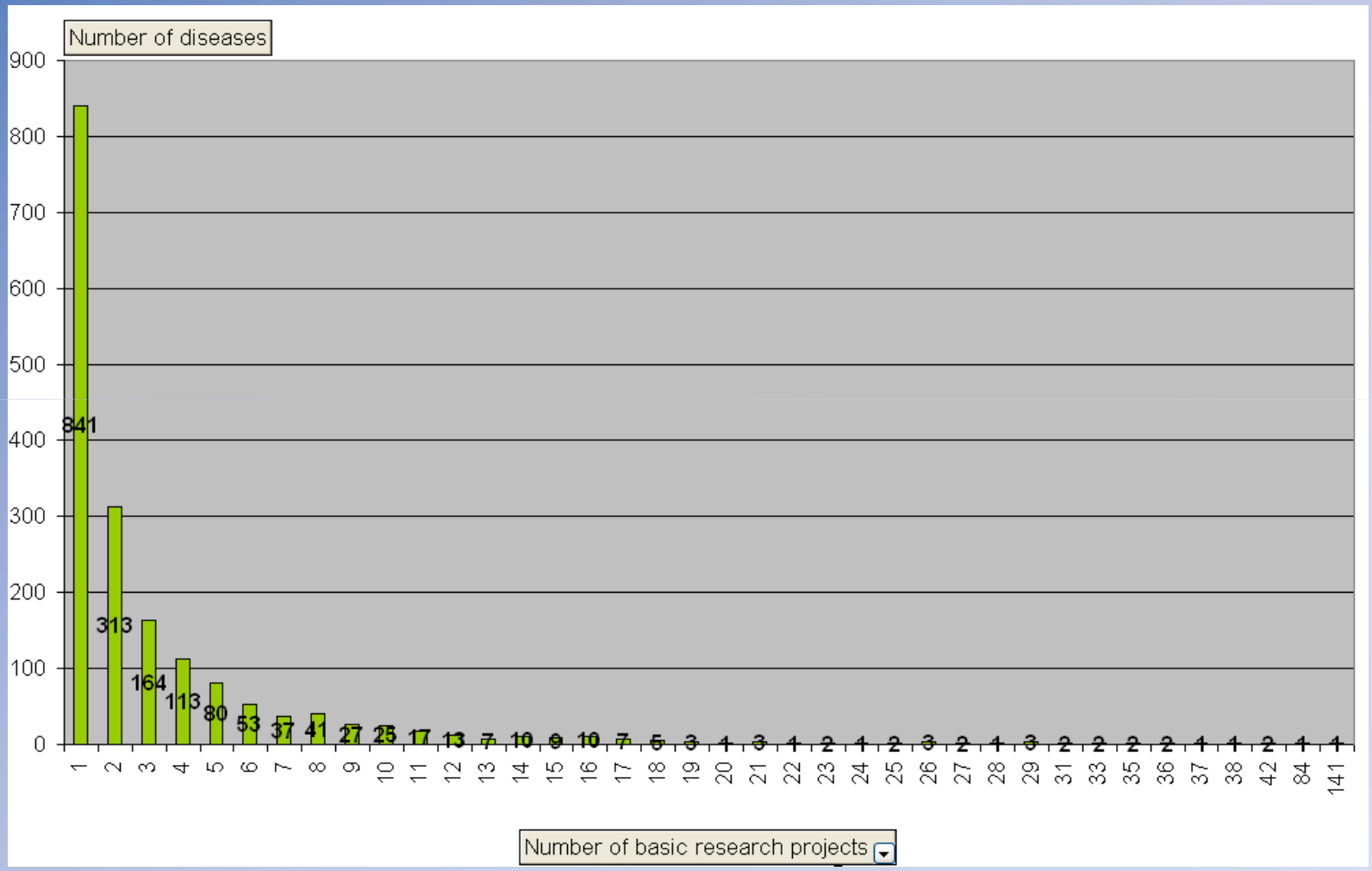
RDPlatform is a FP7 project :

- To provide an overview of the ongoing research on rare diseases in Europe
- To give access to the information on on-going research activities in 25 countries via Orphanet
- To identify new avenues for research
- To describe research infrastructures specific to the RD research
- To provide recommendations to target calls for proposals at the EC and member state level

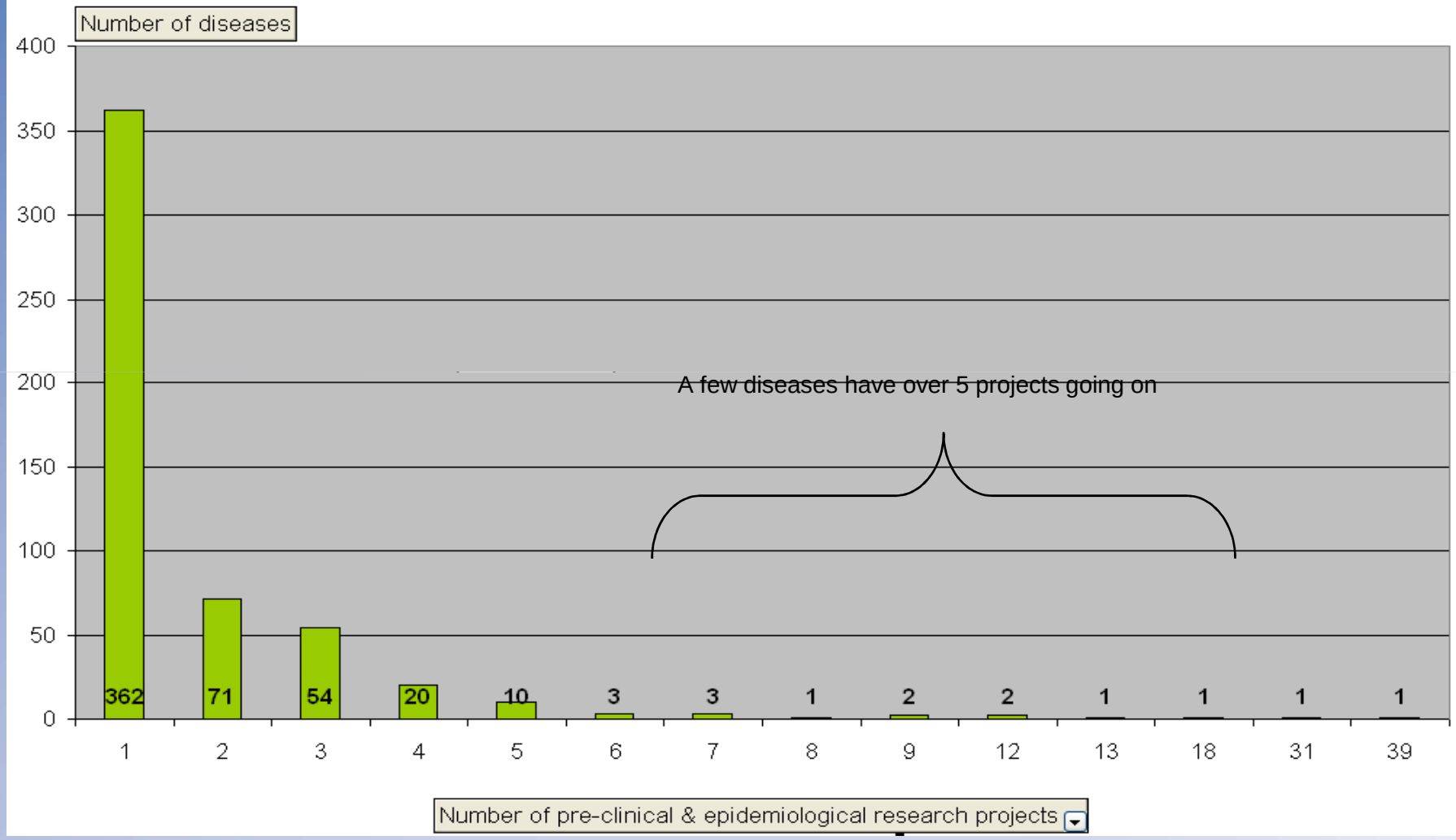
Research in the field of RD: where to we stand ?

- 4 770 ongoing research projects, excluding clinical trials
  - Covering 2121 diseases
  - 524 Gene search
  - 701 Mutation search
  - 255 Gene expression profile
  - 346 Animal model creation/study
  - 353 Genotype/phenotype correlation
  - 89 Biomarker development
  - 313 Diagnostic tool/protocol development
  - 228 Epidemiological studies
  - 174 Observational clinical studies
  - 57 Preclinical cell therapy
  - 121 Preclinical gene therapy
  - 128 Preclinical drug therapy

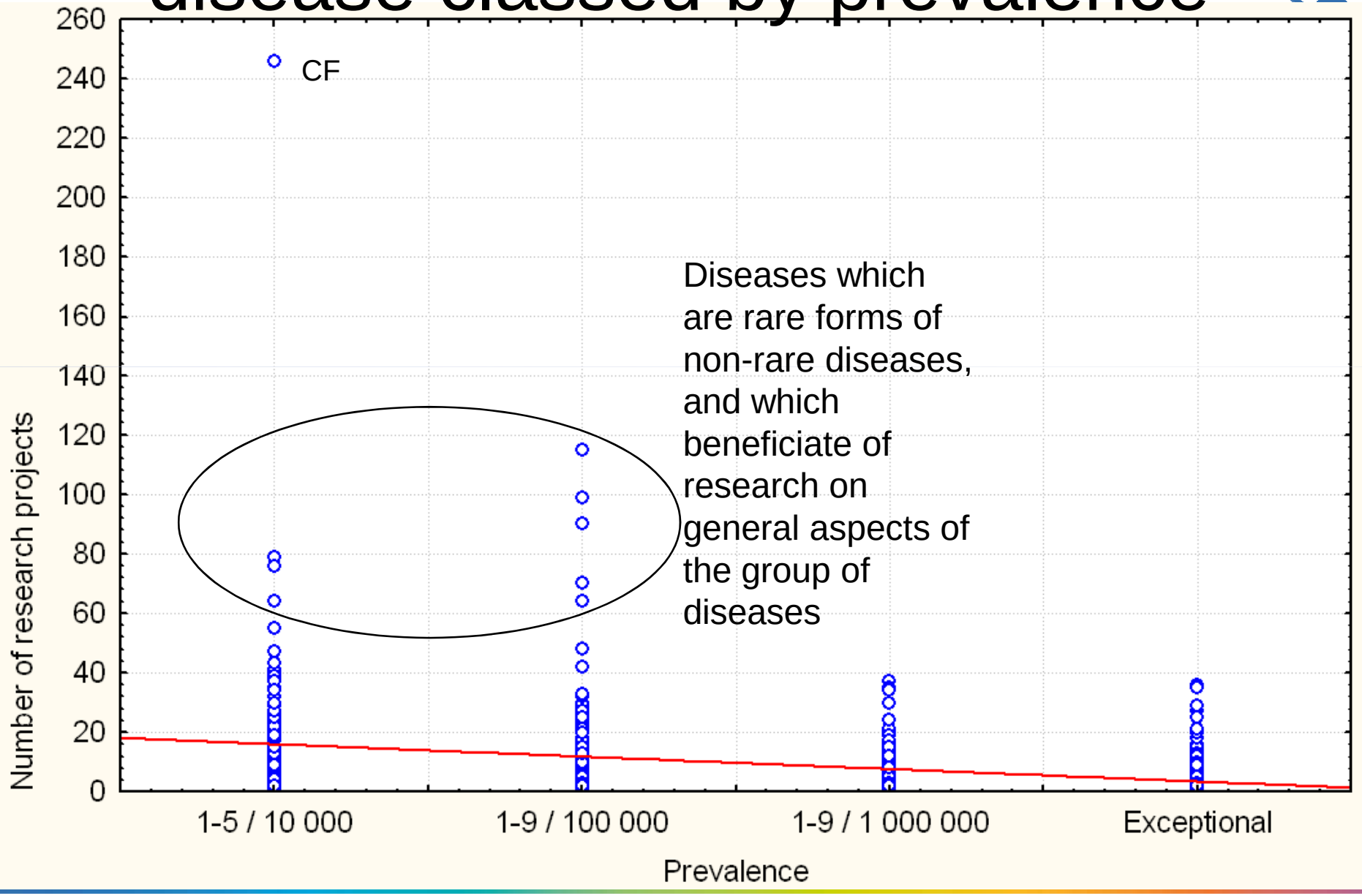
Distribution of number of diseases by number of basic research projects



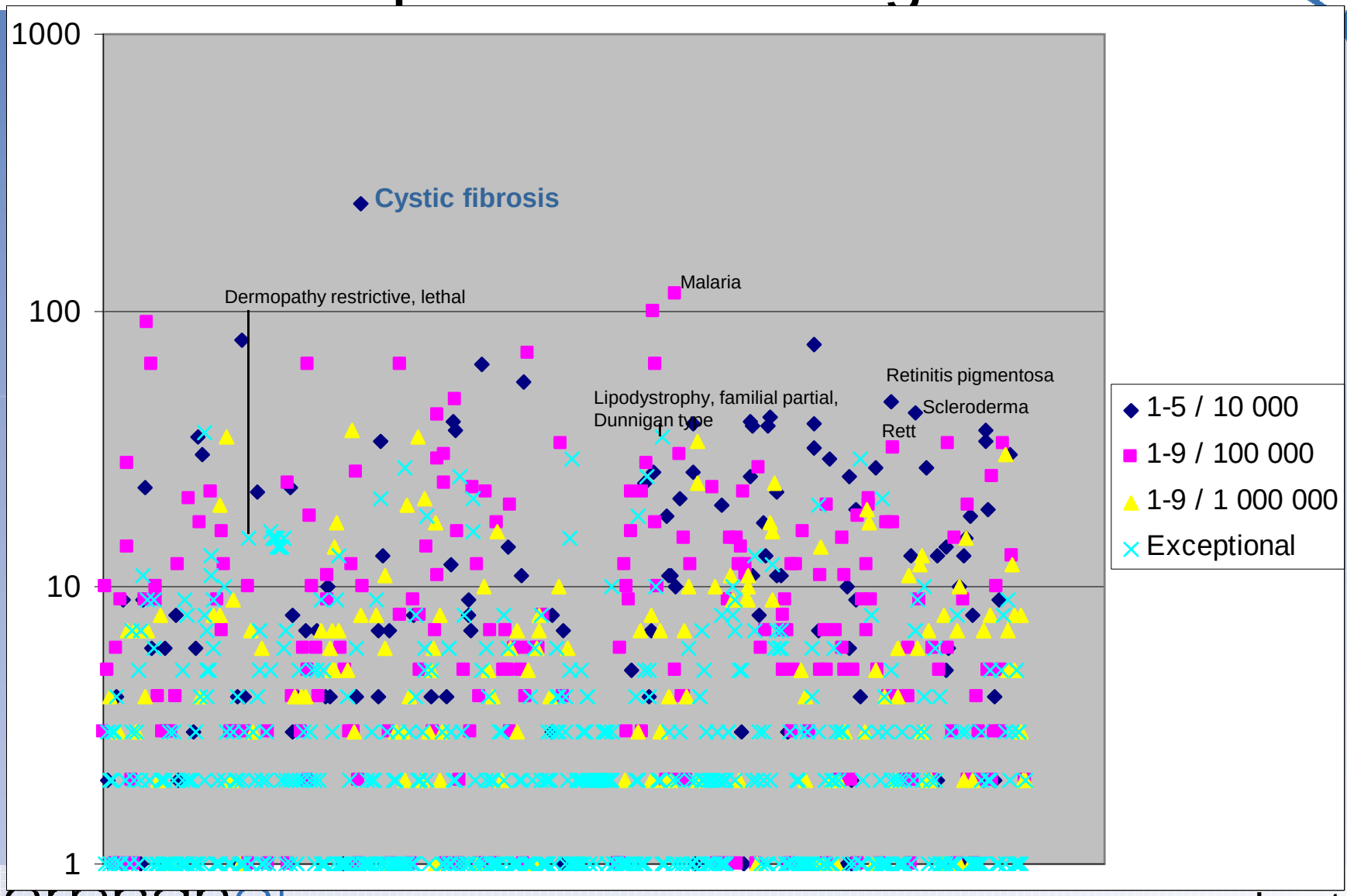
Distribution of number of diseases by number of pre-clinical and epidemiological research projects

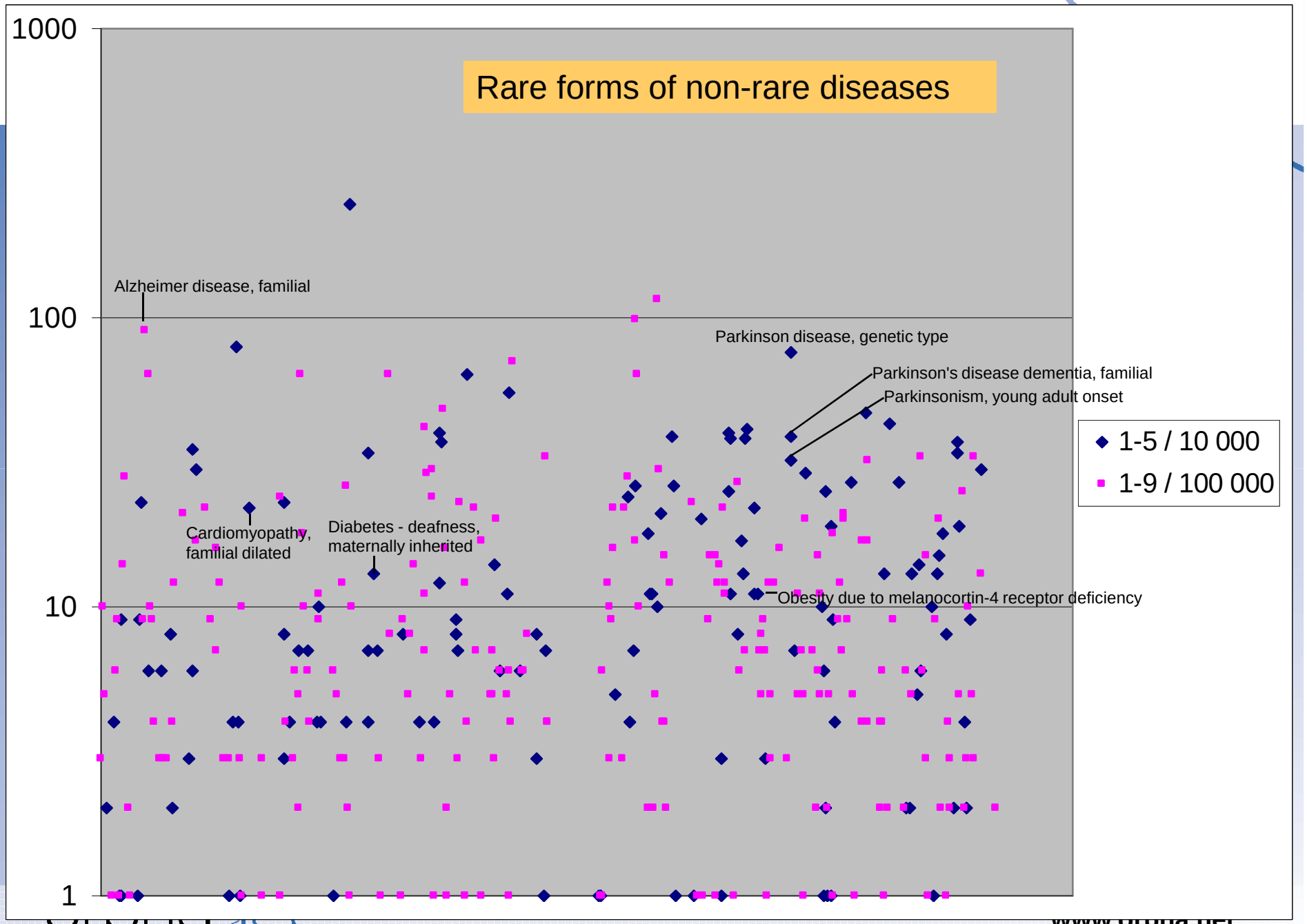


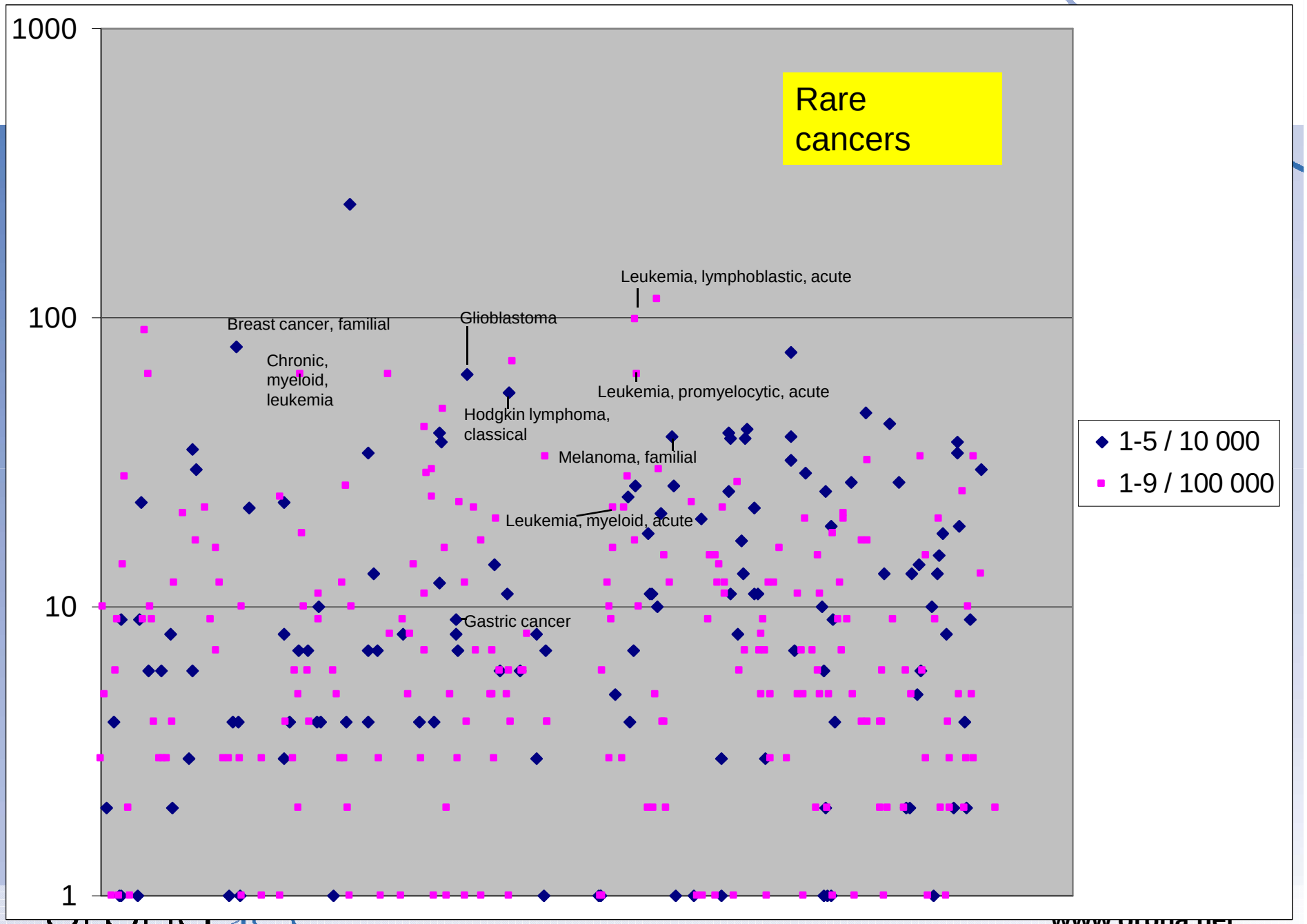
Number of research projects by disease classed by prevalence

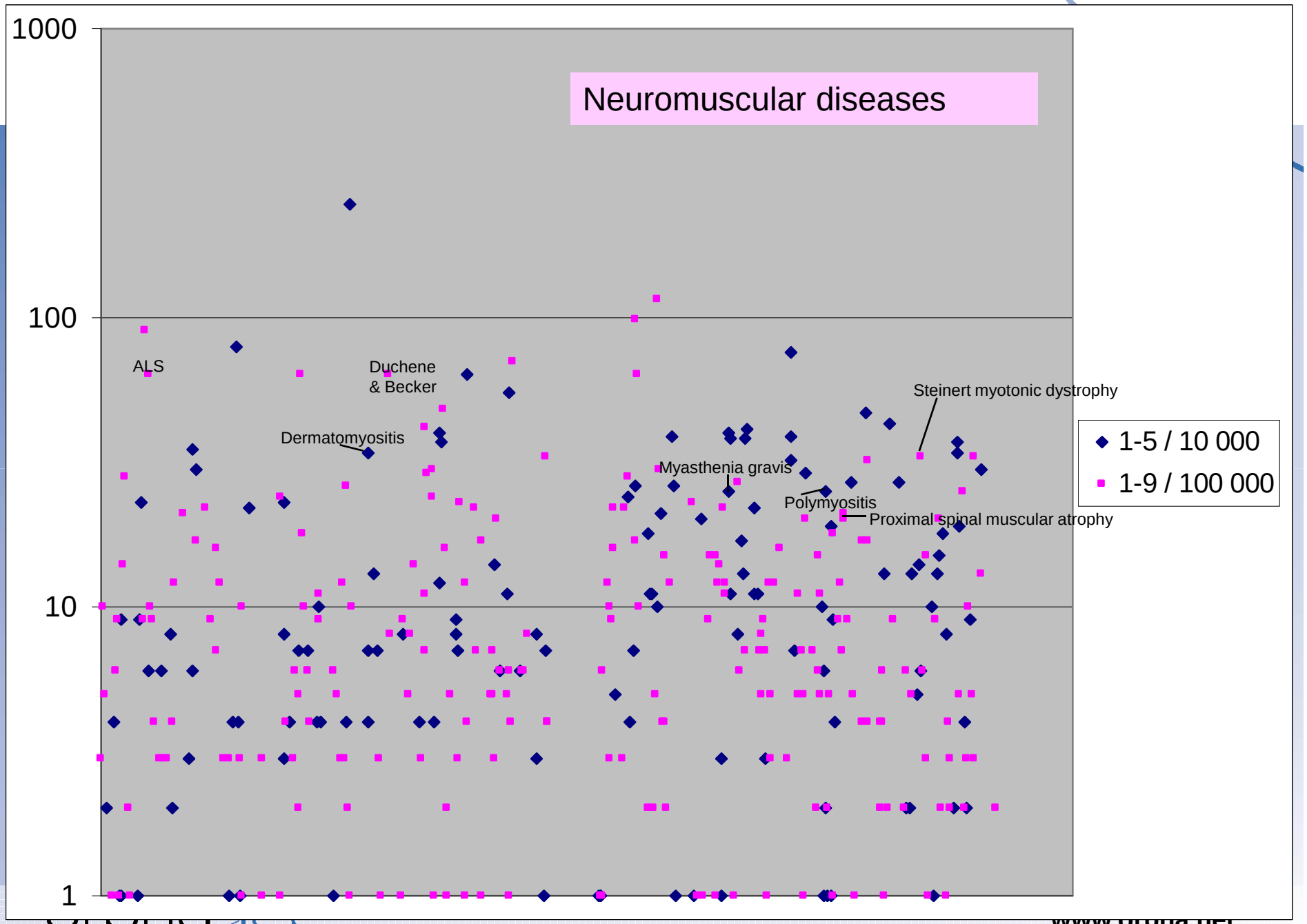


Number of projects by disease by prevalence range



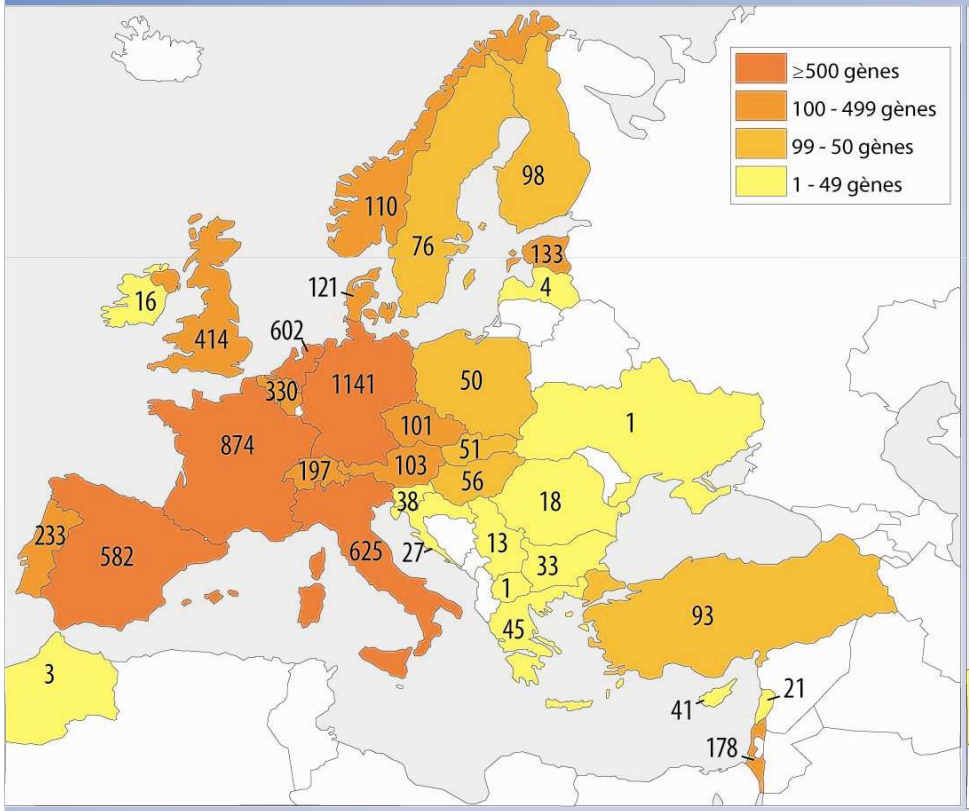

Rare forms of non-rare diseases
1000
100
10
1
Alzheimer disease, familial
Cardiomyopathy, familial dilated
Diabetes - deafness, maternally inherited
Parkinson disease, genetic type
Parkinson's disease dementia, familial
Parkinsonism, young adult onset
Obesity due to melanocortin-4 receptor deficiency
1-5 / 10 000
1-9 / 100 000


Rare cancers
1000
100
10
1
Leukemia, lymphoblastic, acute
Breast cancer, familial
Glioblastoma
Chronic, myeloid, leukemia
Leukemia, promyelocytic, acute
Hodgkin lymphoma, classical
Melanoma, familial
Leukemia, myeloid, acute
Gastric cancer
1-5 / 10 000
1-9 / 100 000

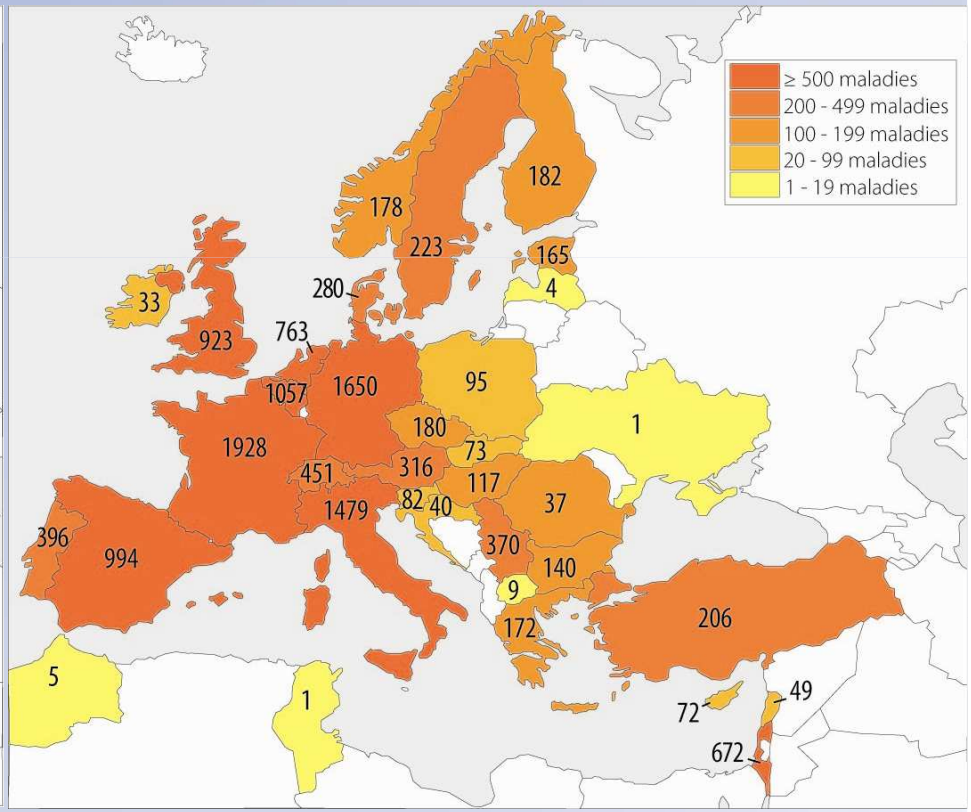

Neuromuscular diseases
1000
100
10
1
ALS
Duchene
& Becker
Dermatomyositis
Myasthenia gravis
Polymyositis
Steinert myotonic dystrophy
Proximal spinal muscular atrophy
1-5 / 10 000
1-9 / 100 000

Major progresses in gene identification translating into diagnostic tests

Number of genes tested by country



Number of diseases tested by country

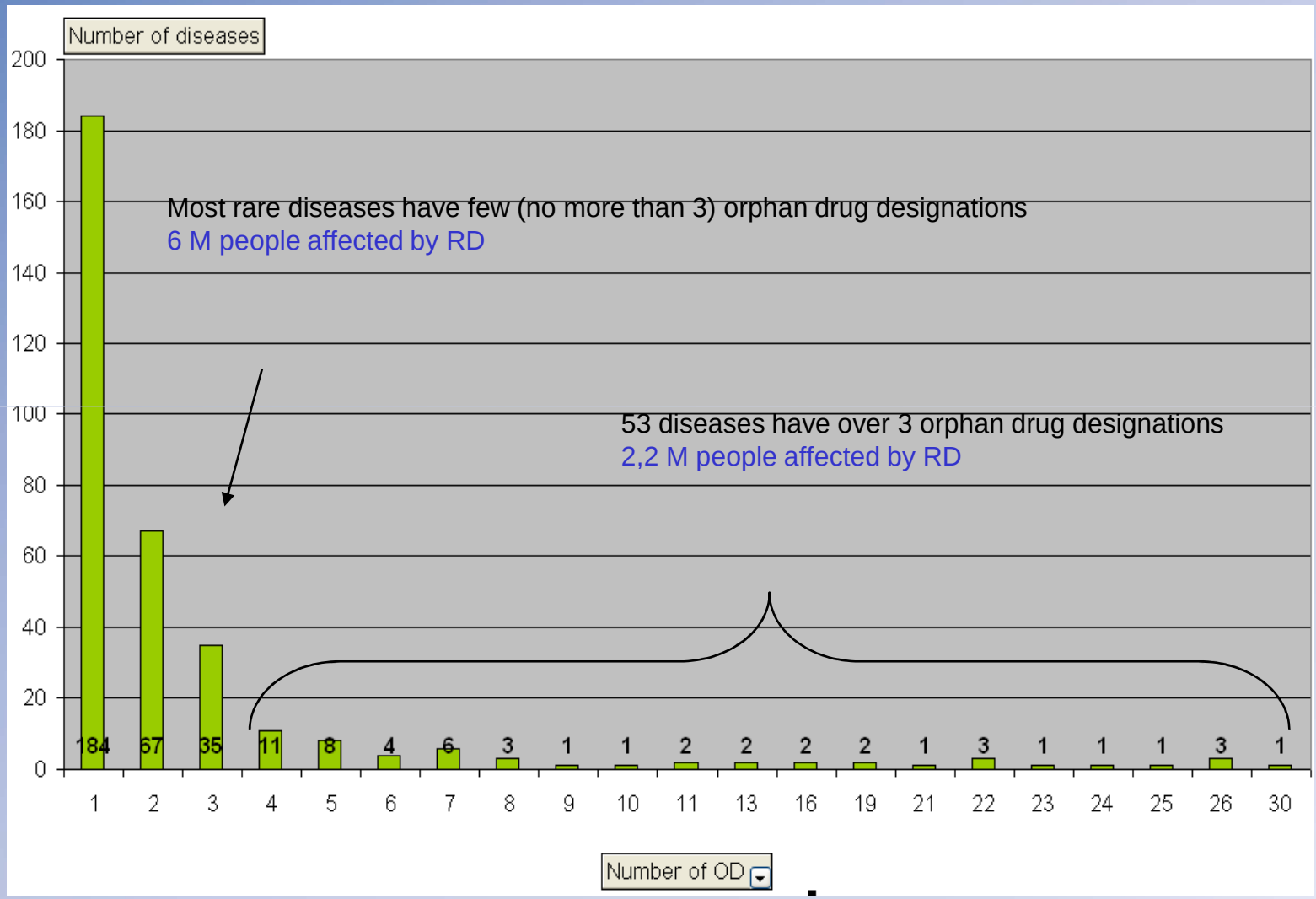


R&D in the field of RD: where do we stand ?

- Potential products in development : Orphan designations as a proxy
  - 581 orphan designations
  - for potentially treating 343 diseases
  - affecting 8.2 Million people
- Products in development: On-going clinical trials
  - 666 ongoing unique trials
  - for potentially 312 diseases
  - affecting 6.8 Million people
- Products on the market:
  - 99 marketed drugs for treating 141 diseases
    - ✓55 drugs with MA and OD in Europe for 82 diseases
    - ✓44 drugs with MA but no OD for 74 diseases

Trends in Orphan Designations

581 Orphan designations are concentrated on 343 rare diseases

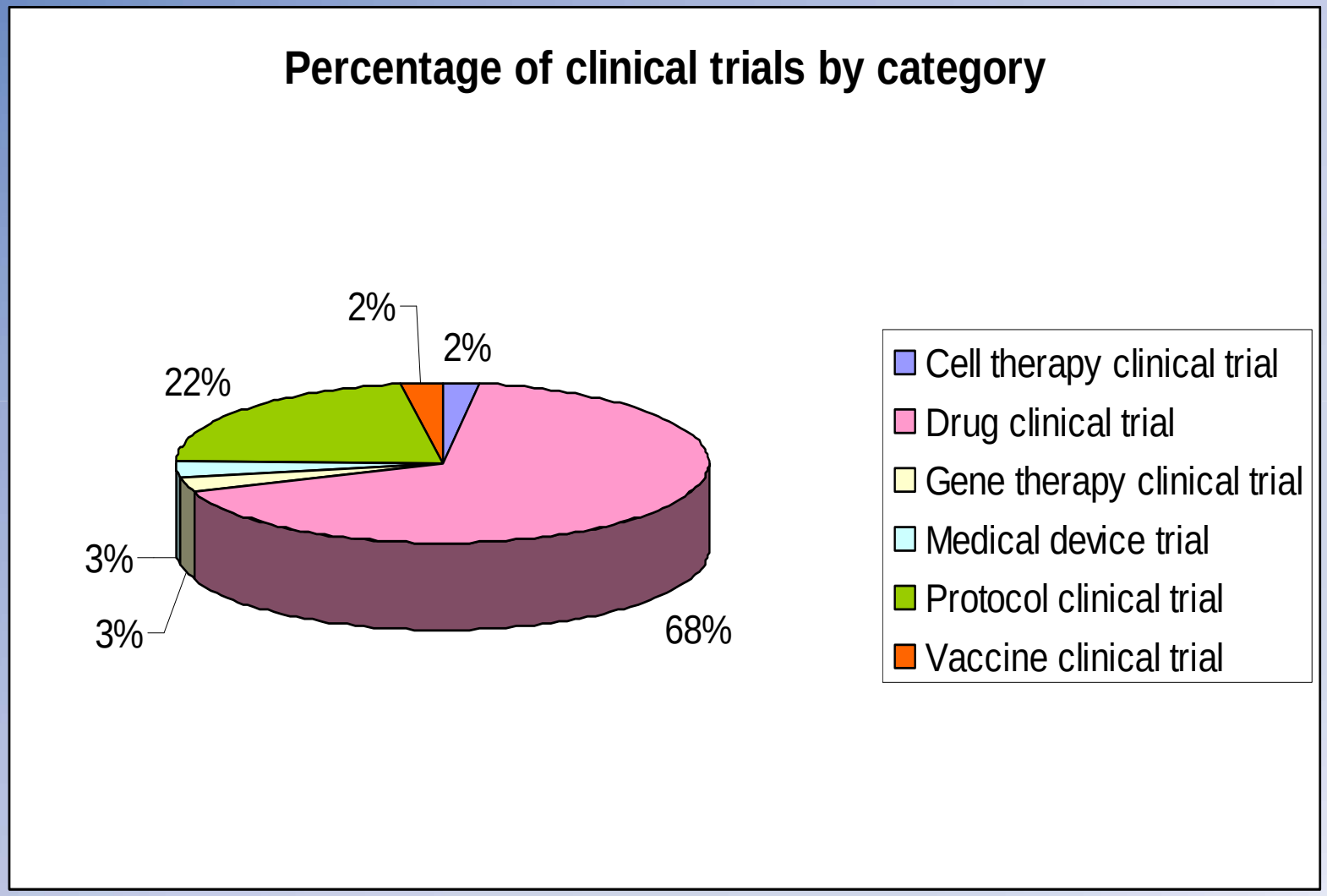


List of non-tumours RD with over 3 orphan designations

- Cystic Fibrosis 30
- Pulm. Art. hypertension 13
- Chronic pulm. hypertension 11
- Graft versus host disease 9
- Acute respiratory distress 7
- Amyotrophic lateral sclerosis 7
- Spinal cord injury 7
- Alpha-1 antitrypsin deficiency 6
- Pulmonary fibrosis, idiopathic 5
- Retinitis pigmentosa 5
- Sickle cell anaemia 5
- Tuberculosis 5
- Beta-thalassemia 4
- Haematopoietic cell transpl. 4
- Duchenne musc. dystrophy 4
- Rejection corneal transplant. 4
- Scleroderma 4

Trends in Clinical Trials

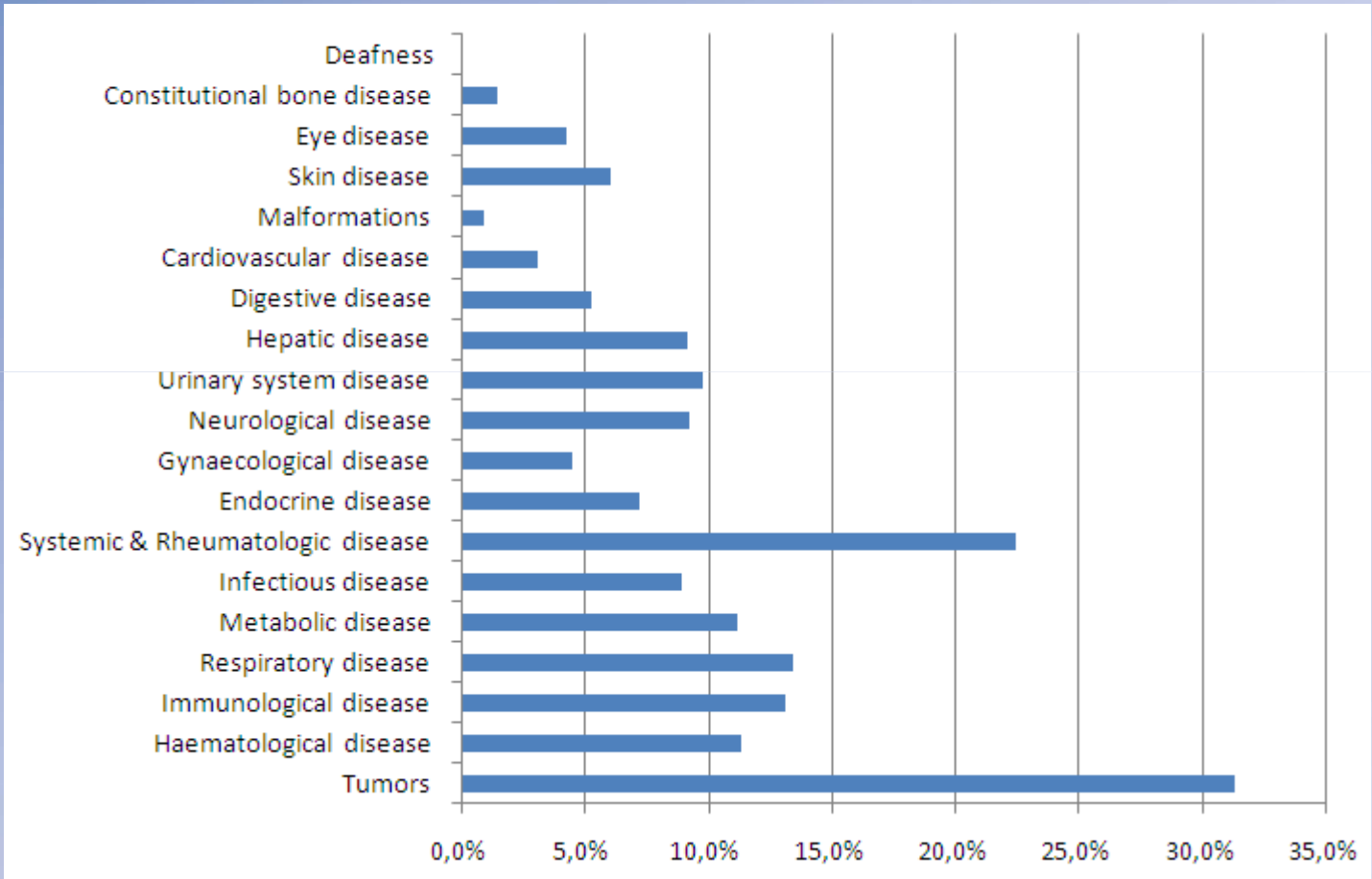
666 ongoing national or international unique clinical trials for 312 diseases



Rare Disease with the highest number of clinical trials in Europe

Leukemia, myeloid, acute	45
Leukemia, lymphoblastic, acute	34
Glioblastoma	32
Myelodysplastic syndromes	28
Cystic fibrosis	27
Atypical hemolytic uremic syndrome	26
Diffuse large B-cell lymphoma	20
Chronic myeloid leukemia	19
Astrocytoma	17
Graft versus host disease	17
Hodgkin lymphoma, classical	17
Myeloma, multiple	17
Friedreich ataxia	16
Mantle cell lymphoma	16
Ependymoma	14
Leukemia, B-cell lymphocytic, chronic	14
Pulmonary fibrosis, idiopathic	14
Follicular lymphoma	13
Leukemia, promyelocytic, acute	13
Amyotrophic lateral sclerosis	12

% of diseases by medical area with at least one product in development (OD &CT)



Drug therapy: R&D by Medical Area
Number of products in development or with MA

Solid tumors	173	Ophthalmology	20
Neurology	151	Bone diseases	17
Haematology	118	Malformations	13
Metabolism	68	Gynecology	13
Dermatology	59	Psychiatry	13
Endocrinology	51	Hepatology	13
Rheumatology	38	Cardiology	10
Immunology	35	Vascular diseases	9
Infectiology	33	Intoxications	7
Gastroenterology	32	Allergology	6
Nephrology	27	Urology	2
Pneumology	23	Maxillofacial	2

Possible Determinants

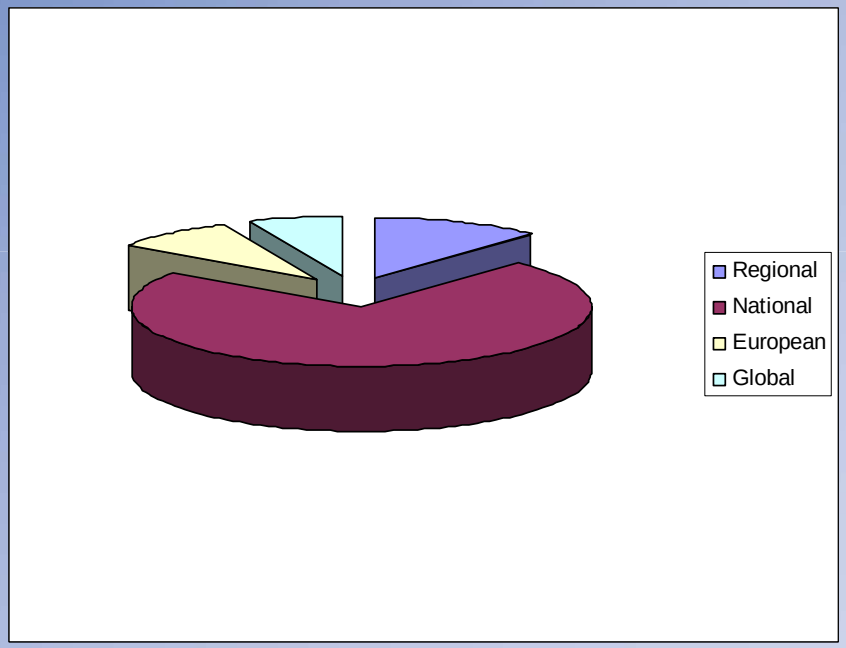
395 Registries as strategic tools

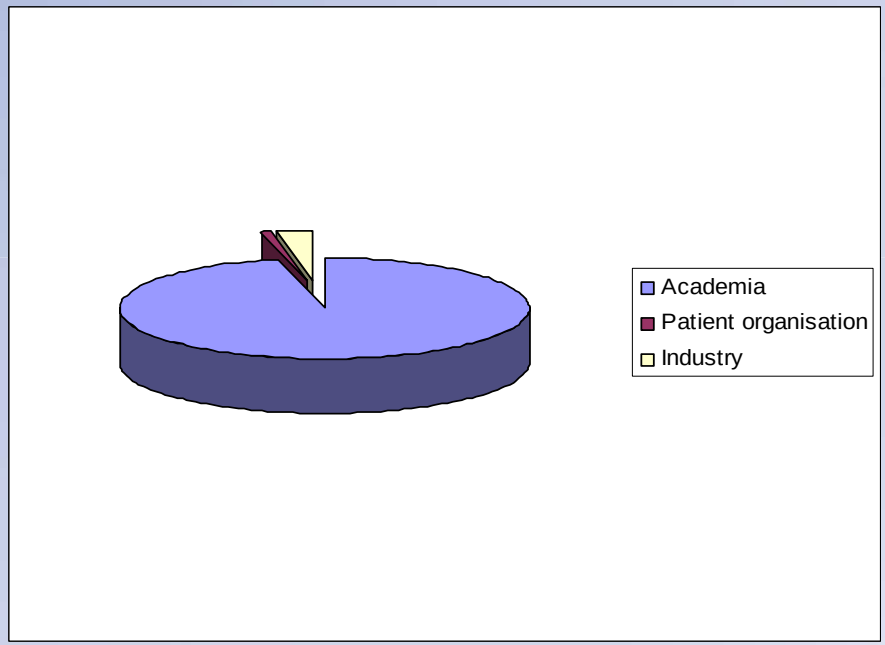
Number of patient registries per country

France	103
Germany	51
Great-Britain	50
Italy	47
Spain	28
Belgium	19
Netherlands	10
Austria	13
Ireland	9
Portugal	7
Switzerland	6
Greece	2
Bulgaria	4
Denmark	3
Romania	2

- Orphanet Report Series on Orphanet front page

Characteristics of Registries



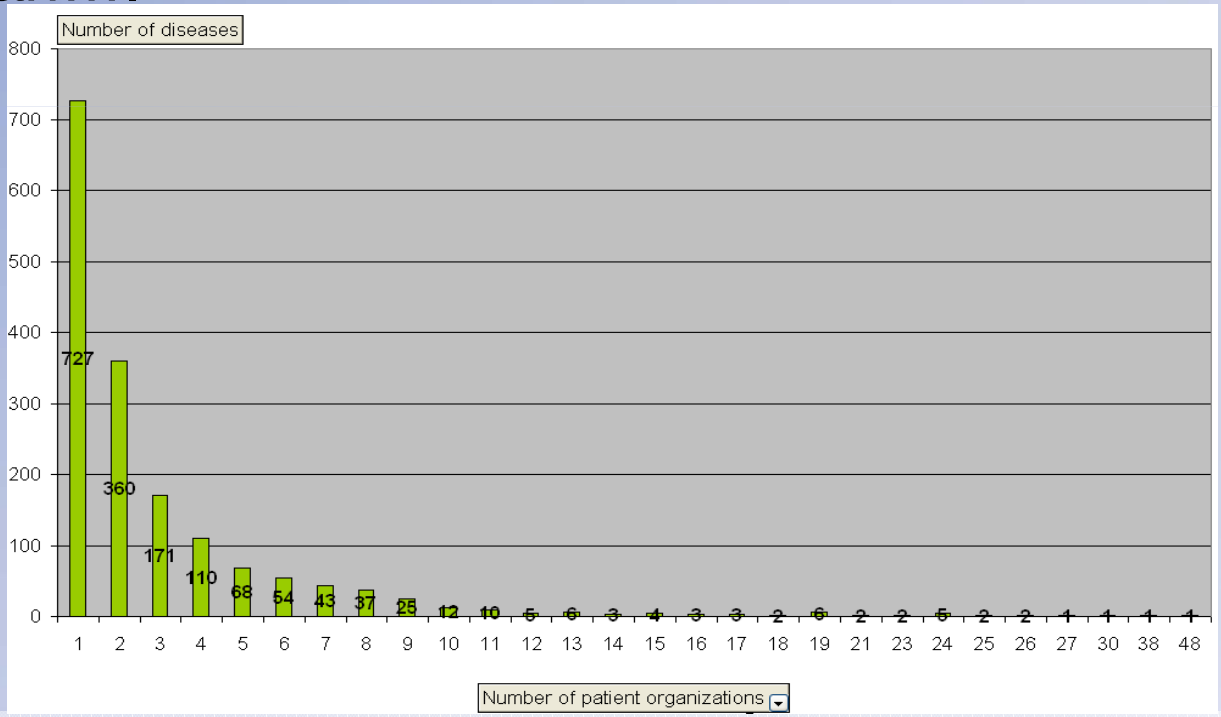


60 International Patient Registries Around a medicinal product

- Cystic fibrosis
- Alpha 1 anti-trypsin
- Bleeding disorders
- Langerhans cell histiocytosis
- Severe chronic neutropenia
- Biliary atresia
- Neuromuscular diseases
- Wilson disease
- Fanconi anemia
- Pulmonary hypertension
- Metabolic diseases: Gaucher, Fabry, Pompe, MPS1…
- Ondine syndrome
- Primary immunodeficiencies
- Retinal dystrophies
- Huntington disease

Distribution of number of diseases covered by 1859 patient organisations (1 667 diseases)

- Most represented: Cystic Fibrosis, neuro-muscular diseases, Fra X, Prader-Willi, Haemophilia, Turner, Scleroderma, Williams, Osteogenesis imperfecta, Marfan….



Conclusion

- No totally neglected areas
- No clue = no research
- Basic research goes on independantly from prevalence
- Major achievements in gene identification translating into tests
- Development in traditional areas for drug therapy + Additional Innovative approach with gene and cell therapy
- Registries, European networks and patient organisations are determinants of R&D
- Research requires a community of interested-parties to reach the critical mass

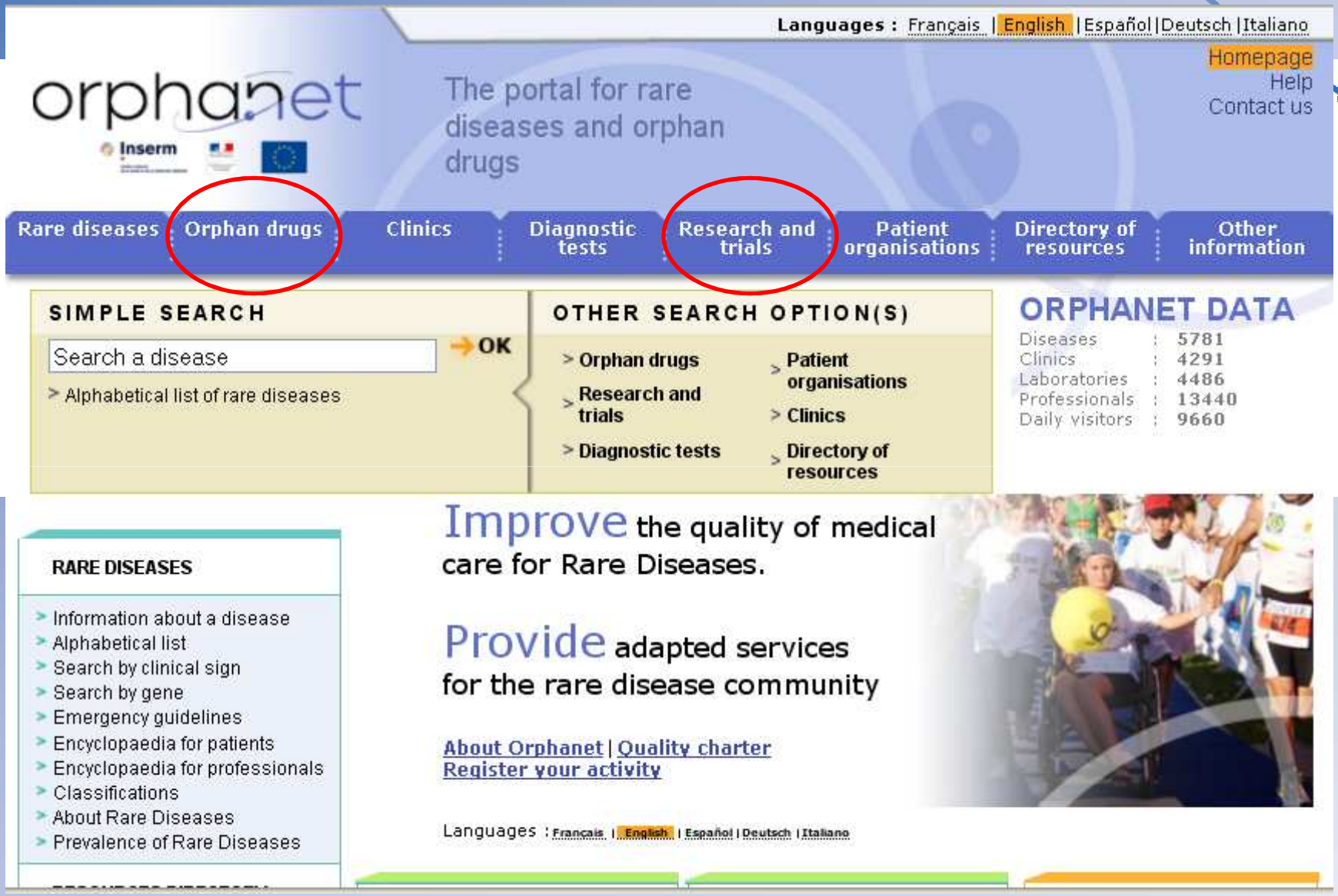
Conclusion

- Disease-specific factors associated with Drug development
  (Drug Discovery Today Oct 2009 by Heemstra et al.)
- Prevalence: significant difference for > and < to 1 in 10,000
- Disease class: traditional classes as research is a process
- Disease-specific scientific output:
  - ✓Diseases with more than 600 published papers have twofold higher likelihood to have a product compared to diseases with 200 papers

Conclusion (2)

- Need for Public funding for:
  - To go from hypothesis to proof of concept and first clinical evidence
  - To establish and maintain registries
  - To run academic clinical trials for orphan indications
  - To run protocol trials
- Need for public/private partnership for Registries
- Need to bridge the gap between basic research and clinical research
  - develop databases, information systems, biobanks in general
  - Invest on Web 3.0, the semantic web
- Need to regularly organise surveys about possible valuable hypothesis, not tested for reasons possible to address

Many facilities to retrieve information on research activities, clinical trials and orphan drugs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Alphabetical list of rare diseases
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ORPHANET DATA
Diseases : 5781
Clinics : 4291
Laboratories : 4486
Professionals : 13440
Daily visitors : 9660
RARE DISEASES
Information about a disease
Alphabetical list
Search by clinical sign
Search by gene
Emergency guidelines
Encyclopaedia for patients
Encyclopaedia for professionals
Classifications
About Rare Diseases
Prevalence of Rare Diseases
Improve the quality of medical care for Rare Diseases.
Provide adapted services for the rare disease community
About Orphanet | Quality charter
Register your activity
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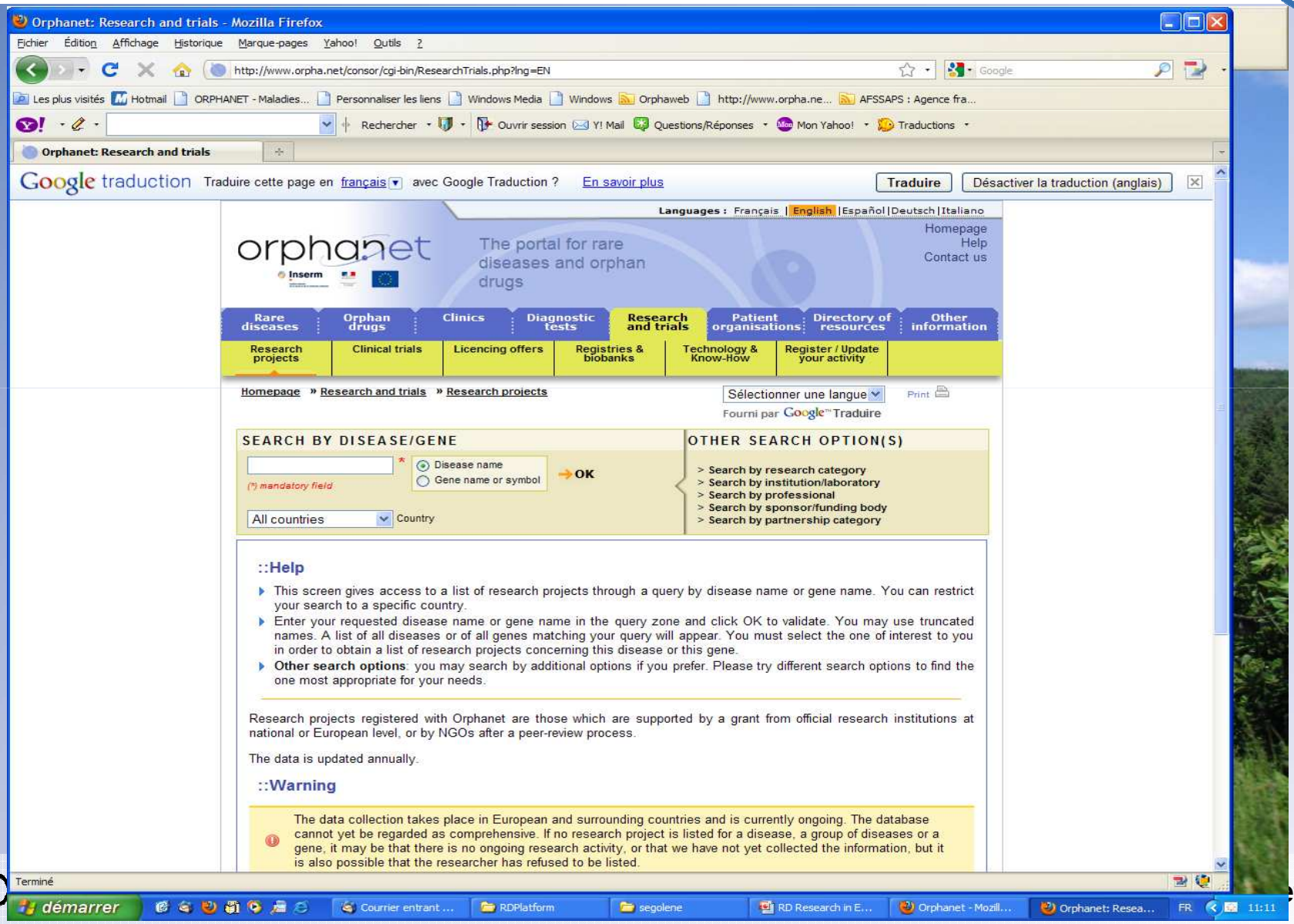
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> Search by institution/laboratory
> Search by professional
> Search by sponsor/funding body
> Search by partnership category
::Help
This screen gives access to a list of research projects through a query by disease name or gene name. You can restrict your search to a specific country.
Enter your requested disease name or gene name in the query zone and click OK to validate. You may use truncated names. A list of all diseases or of all genes matching your query will appear. You must select the one of interest to you in order to obtain a list of research projects concerning this disease or this gene.
Other search options: you may search by additional options if you prefer. Please try different search options to find the one most appropriate for your needs.
Research projects registered with Orphanet are those which are supported by a grant from official research institutions at national or European level, or by NGOs after a peer-review process.
The data is updated annually.
::Warning
The data collection takes place in European and surrounding countries and is currently ongoing. The database cannot yet be regarded as comprehensive. If no research project is listed for a disease, a group of diseases or a gene, it may be that there is no ongoing research activity, or that we have not yet collected the information, but it is also possible that the researcher has refused to be listed.
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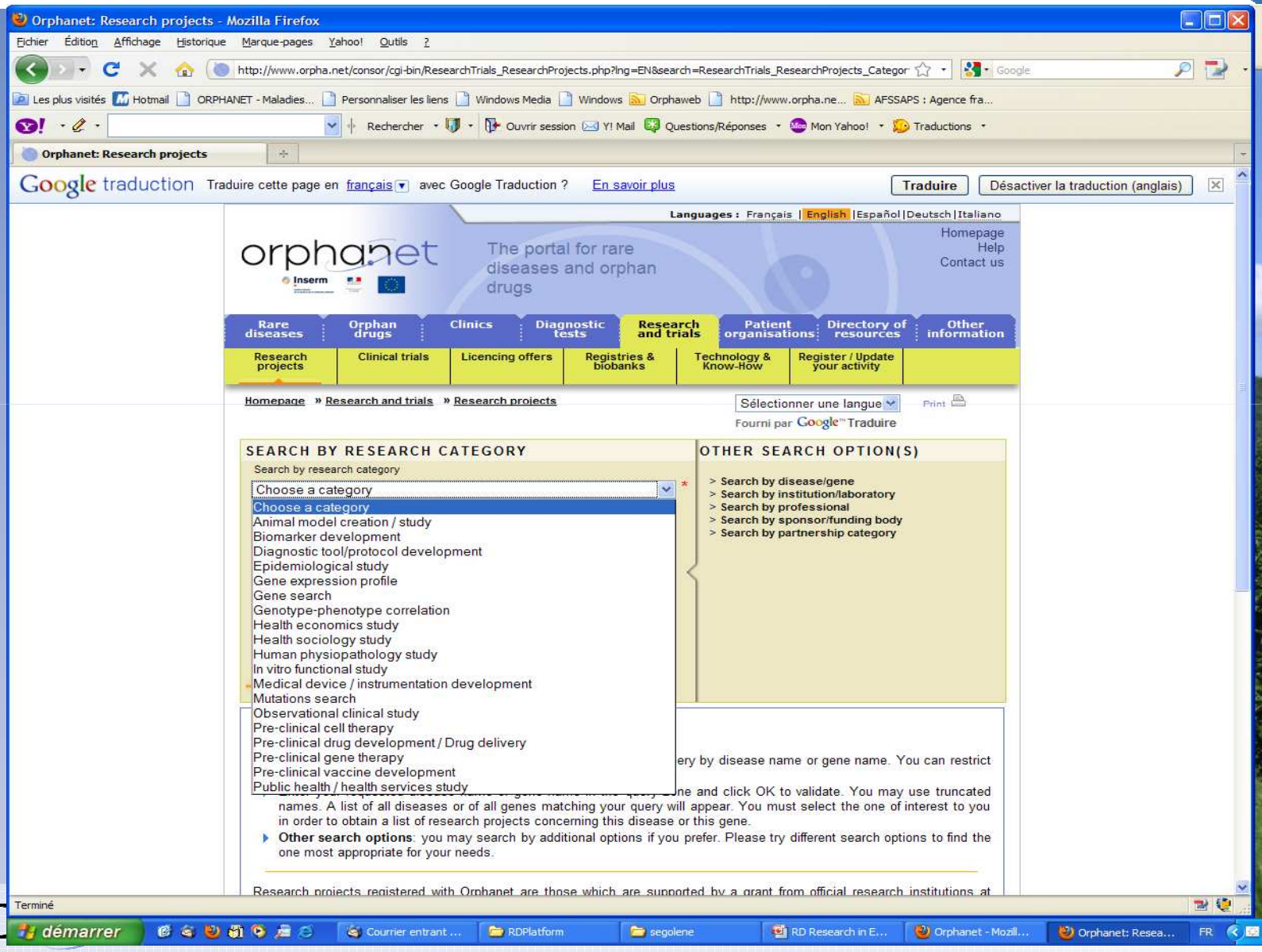
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Health sociology study
Human physiopathology study
In vitro functional study
Medical device / instrumentation development
Mutations search
Observational clinical study
Pre-clinical cell therapy
Pre-clinical drug development / Drug delivery
Pre-clinical gene therapy
Pre-clinical vaccine development
Public health / health services study
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> Search by institution/laboratory
> Search by professional
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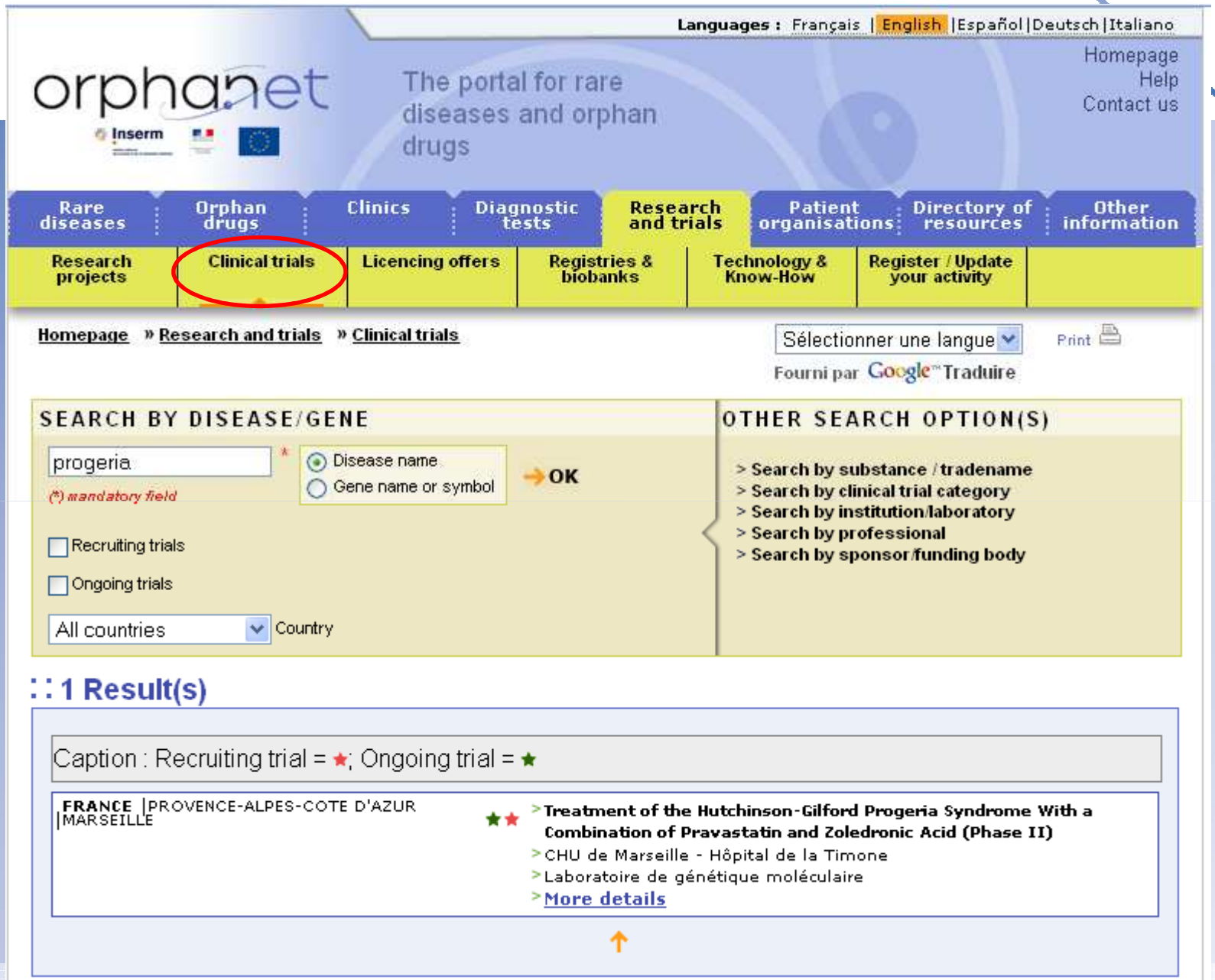
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Ongoing trials
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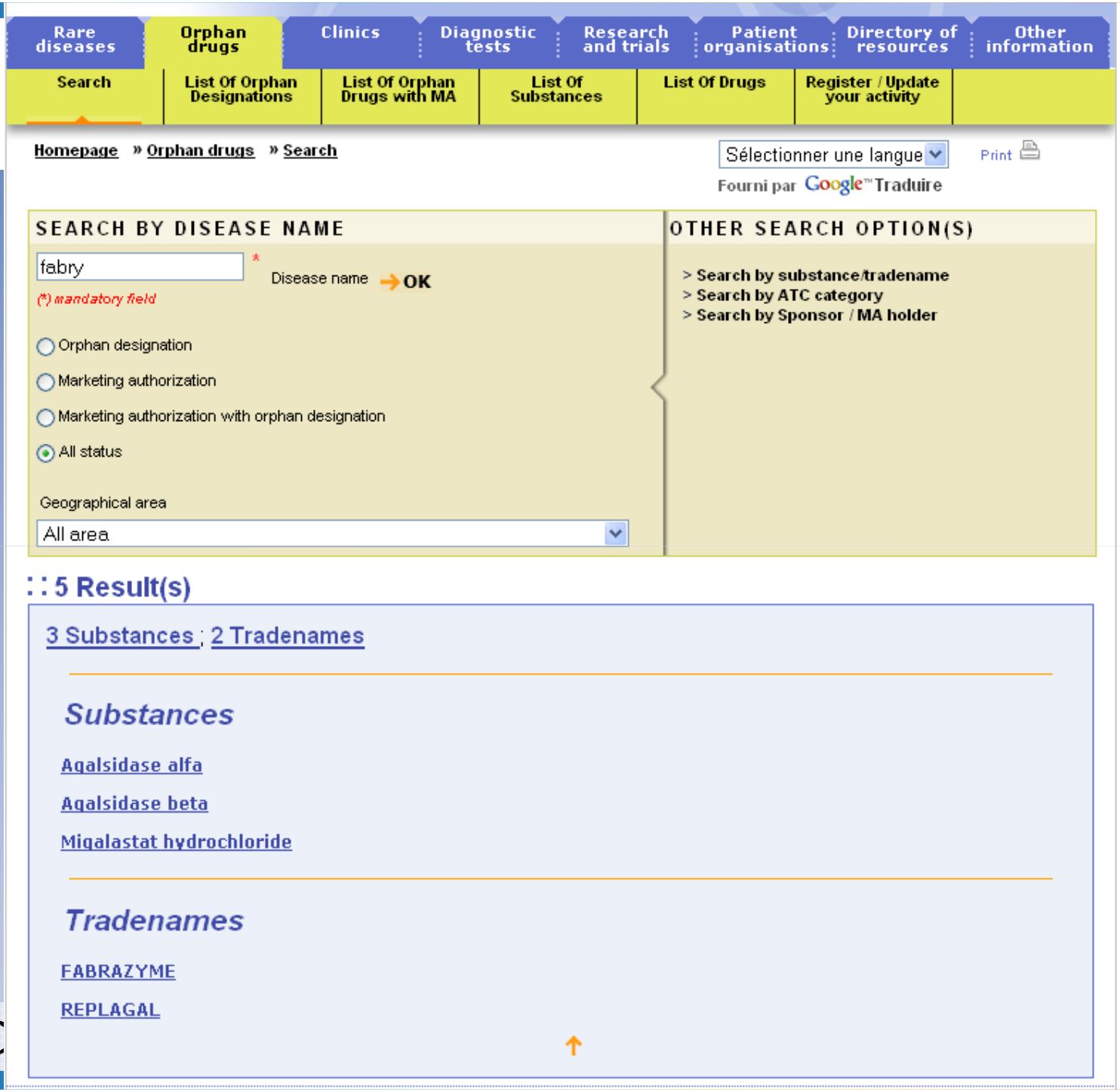
> Search by institution/laboratory
> Search by professional
> Search by sponsor/funding body
: : 1 Result(s)
Caption : Recruiting trial = ★; Ongoing trial = ★
FRANCE | PROVENCE-ALPES-COTE D'AZUR | MARSEILLE
★★
> Treatment of the Hutchinson-Gilford Progeria Syndrome With a Combination of Pravastatin and Zoledronic Acid (Phase II)
> CHU de Marseille - Hôpital de la Timone
> Laboratoire de génétique moléculaire
> More details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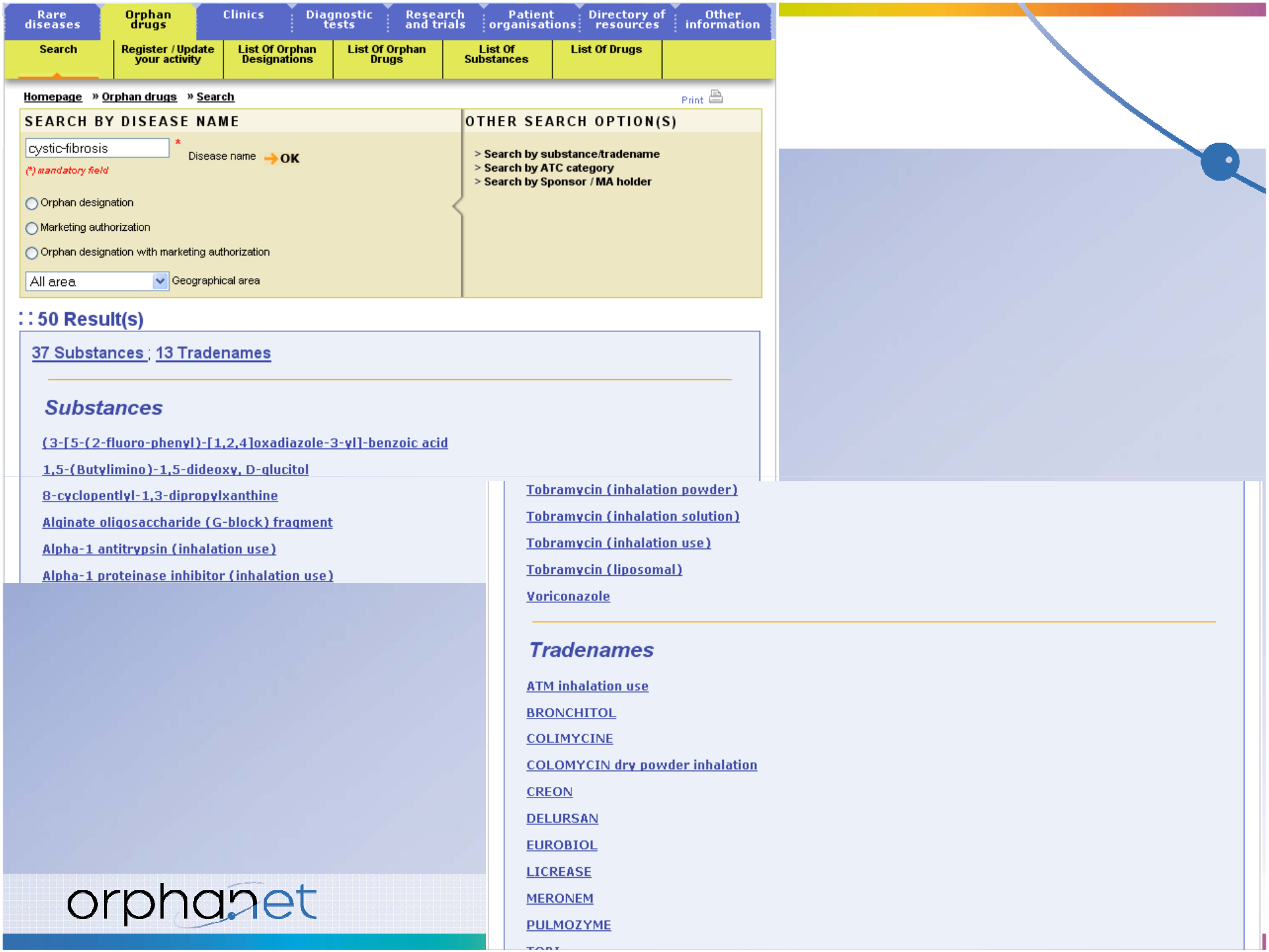
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8-cyclopentlyl-1,3-dipropylxanthine
Alginate oligosaccharide (G-block) fragment
Alpha-1 antitrypsin (inhalation use)
Alpha-1 proteinase inhibitor (inhalation use)
Tobramycin (inhalation powder)
Tobramycin (inhalation solution)
Tobramycin (inhalation use)
Tobramycin (liposomal)
Voriconazole
Tradenames
ATM inhalation use
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COLOMYCIN dry powder inhalation
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DELURSAN
EUROBIOL
LICREASE
MERONEM
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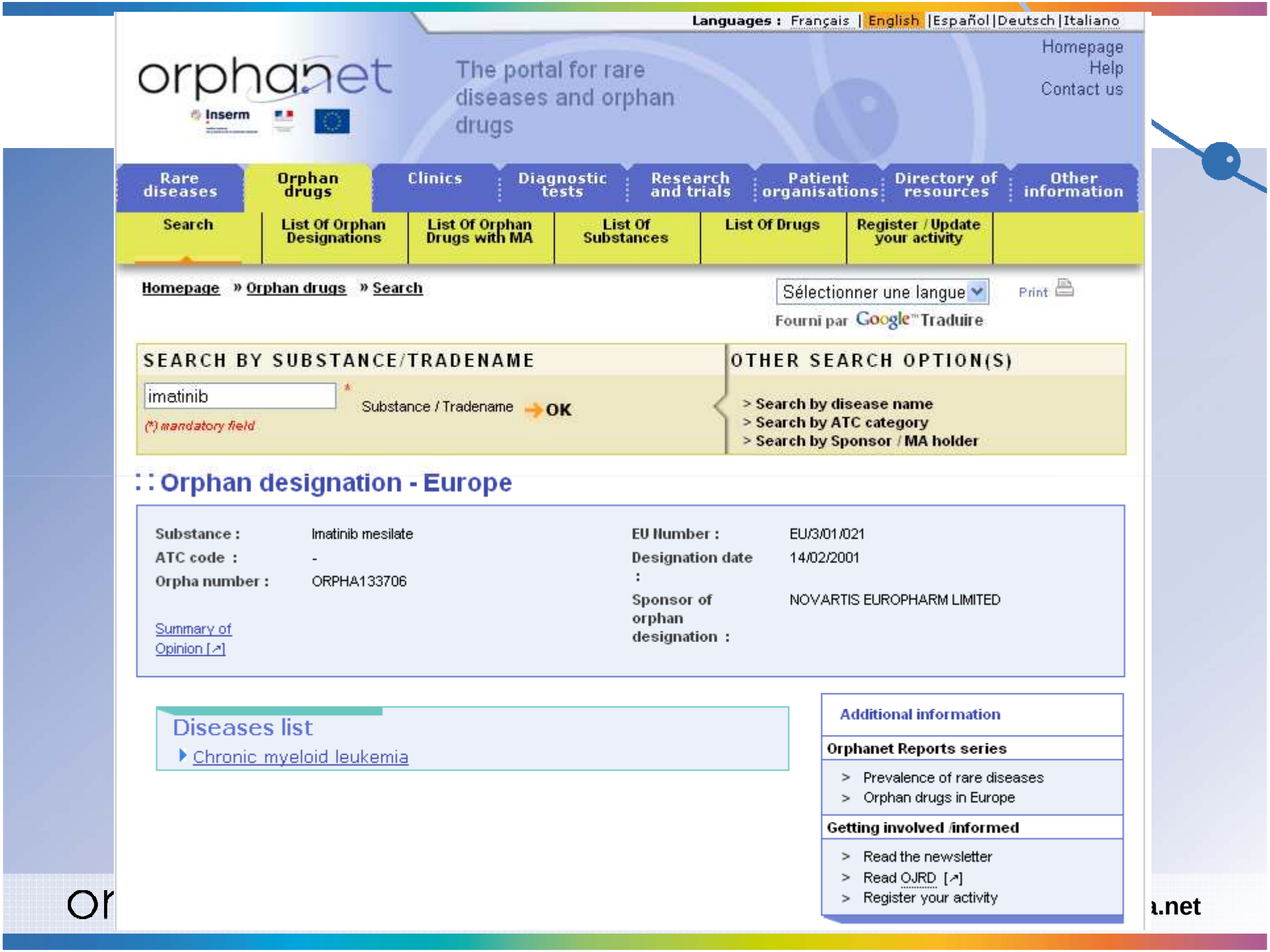
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> Search by ATC category
> Search by Sponsor / MA holder
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Summary of Opinion [↗]
EU Number : EU/3/01/021
Designation date : 14/02/2001
Sponsor of orphan designation : NOVARTIS EUROPHARM LIMITED
Diseases list
Chronic myeloid leukemia
Additional information
Orphanet Reports series
> Prevalence of rare diseases
> Orphan drugs in Europe
Getting involved /informed
> Read the newsletter
> Read OJRD [↗]
> Register your activity

Thank you for your attention