

Rare diseases research in Europe: an overview based on data from the Orphanet database (Nov 2009)

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Objective:

- Provide an overview of ongoing research in Europe (data from funding bodies)
- Identify trends and determinants and possible new avenues
- Provide recommendations to funding bodies concerning topics to address

Methods:

- Analysis of data collected by Orphanet teams in 34 countries
- Data on basic and clinical research, clinical trials, registries, networks, products in development and marketed products
- Analysis performed by medical area according to Orphanet classification

Ongoing basic and clinical research in Europe: 5867 projects identified by Orphanet

- 534 Gene searches
- 713 Mutations searches
- 245 Gene expression profiles
- 328 Animal model creation/ studies
- 966 In vitro functional studies
- 356 Genotype-phenotype correlations
- 868 Human physiopathology studies
- 117 Pre-clinical gene therapies
- 51 Pre-clinical cell therapies
- 117 Pre-clinical drug development
- 30 Pre-clinical vaccine development
- 29 Observational clinical studies
- 25 Medical device/instrumentation development
- 102 Biomarkers development
- 324 Diagnostic tool/protocol development
- 257 Epidemiological studies
- 666 Clinical trials
- 450 Patient registries
- 98 Health sociology studies
- 18 Health economics studies
- 117 Public health/health services studies



R&D in the field of therapy development

Potential products in development: orphan designations as a proxy

- **581 orphan designations**
- to potentially treat **343 diseases**
- affecting **8.2 million people** in Europe

Products in development: ongoing clinical trials

- **666 unique ongoing trials**
- to potentially treat **312 diseases**
- affecting **6.8 million people** in Europe



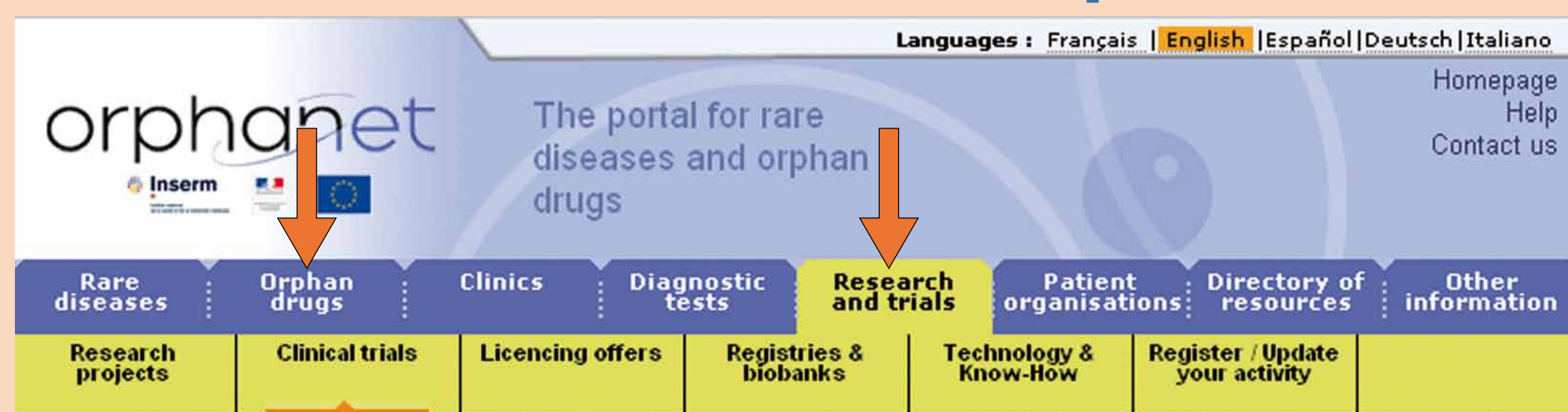
Products on the market in Europe

- **99 marketed drugs** to treat **141 diseases**
- **55 drugs** with MA and OD for **82 diseases**
- **44 drugs** with MA but no OD for **74 diseases**

Drug development: R&D by medical area Number/% of diseases with at least one product in development

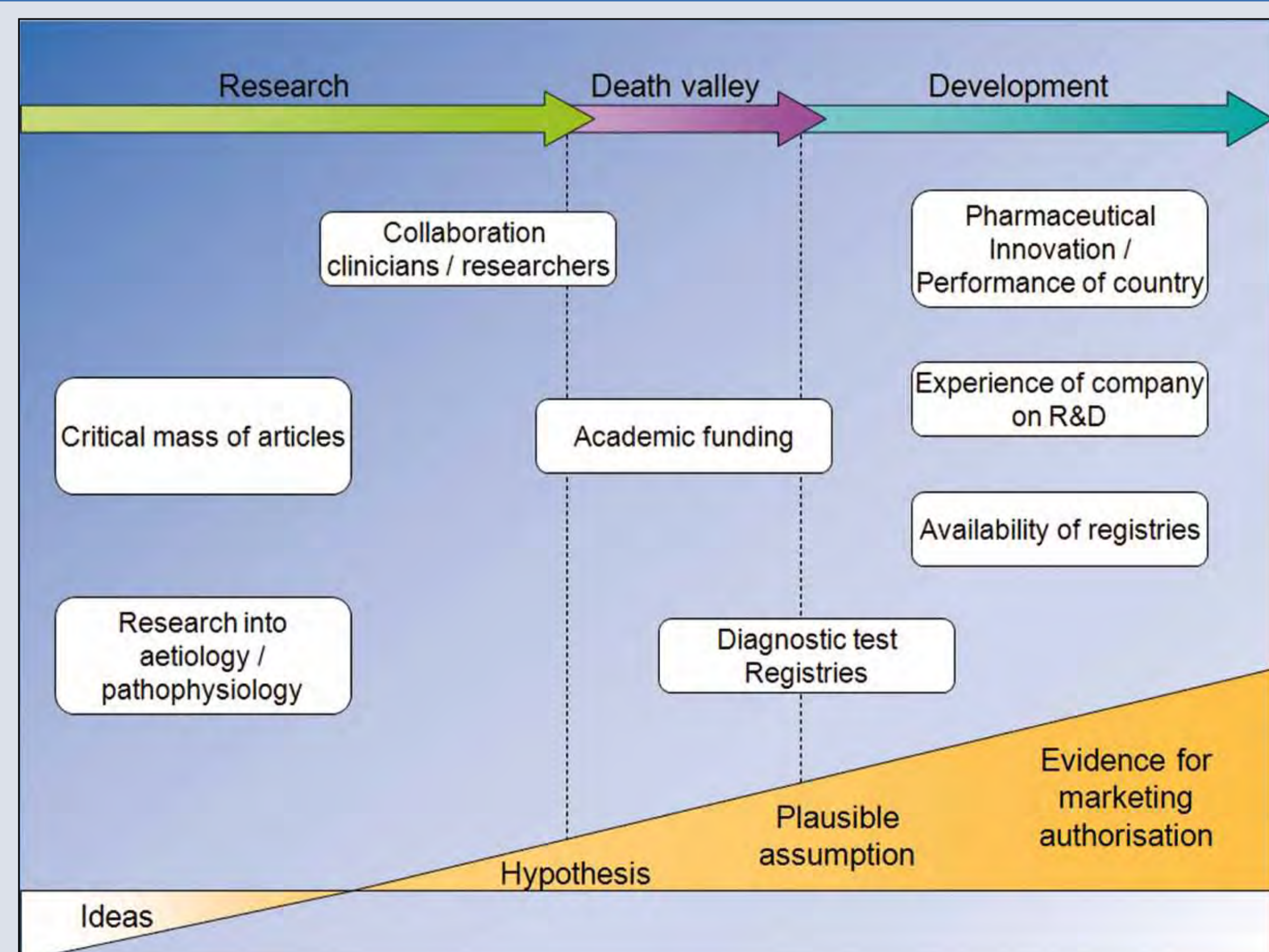
Tumors	152	27.2 %	Respiratory disease	16	2.9 %
Neurological disease	96	17.2 %	Eye disease	16	2.9 %
Metabolic disease	59	10.6 %	Infectious disease	13	2.3 %
Systemic & Rheumatologic disease	31	5.6 %	Cardiovascular disease	9	1.6 %
Endocrine disease	29	5.2 %	Digestive disease	8	1.4 %
Skin disease	26	4.7 %	Hepatic disease	7	1.3 %
Haematological disease	24	4.3 %	Constitutional bone disease	6	1.1 %
Immunological disease	22	3.9 %	Gynaecological disease	5	0.9 %
Urinary system disease	22	3.9 %	Odontological disease	0	0.0 %
Malformations	17	3.0 %	Deafness	0	0.0 %

Access data on www.orpha.net



& www.rdplatform.org

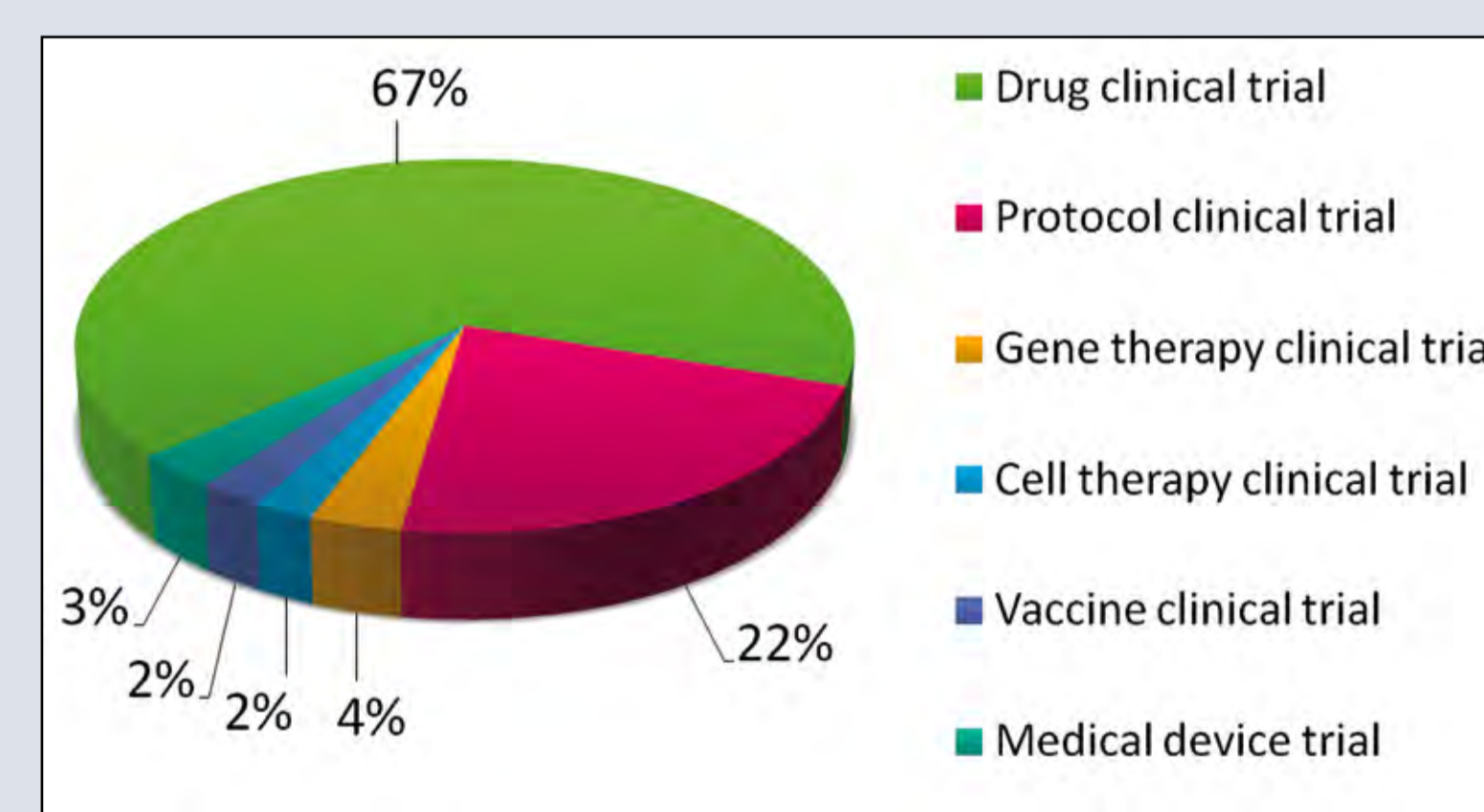
Determinants of research



666 ongoing national or international clinical trials for 312 diseases

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

Rare diseases with the highest number of clinical trials in Europe



Percentage of clinical trials by category

Conclusion:

- No totally neglected medical areas
- Basic research is conducted independent of prevalence
- Development in traditional areas for drug therapy + additional innovative approach with gene and cell therapy
- No clue = no research
- Registries, European networks and patient organisations are determinants of R&D
- Research requires a community of interested parties to reach critical mass

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