ORPHANET’s Services for Researchers

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Database Manager
Orphanet
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<th>Needs</th>
<th>Solutions</th>
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<td>Free-access list</td>
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Indexing rare diseases

Indexing

- clinical signs and symptoms
- OMIM #: catalogue of genes and genetic diseases
- ICD-10 code: international coding system of WHO
- MeSH: Medical Subject Heading
- genes, mode of inheritance
- age group of appearance, prevalence

Access to other documentary resources

- Links to Medline for each rare disease
- Links to reference websites
- Links to a gene database: GenAtlas

Key figures

5,500 diseases
1,125 diseases indexed with MeSH
1,600 diseases with links to Medline
5,205 links to other sites
### List of Current Research Projects

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<th>Genetic research</th>
<th>Development of a new diagnostic protocol</th>
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<td>Gene mutation research</td>
<td>Development of a new medical device</td>
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<td>Physiopathology and model <em>in vitro</em></td>
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<td>Physiopathology and model <em>in vivo</em></td>
<td>Clinical trial of a new protocol</td>
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<td>Natural history</td>
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<td>Gene therapy</td>
<td>Epidemiological studies</td>
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<td>Cell therapy</td>
<td>Registries/ Observatories/ Cohorts</td>
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<tr>
<td>Biomarkers</td>
<td>Epidemiological studies</td>
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<tr>
<td>Biological collections / resources</td>
<td>Research in health sociology</td>
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<td>Preclinical development of a new drug</td>
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<tr>
<td>Development of a new diagnostic instrument</td>
<td>Public Health</td>
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### Key figures

In Europe: **4 625** projects of which **13%** are clinical trials
Collaboration in Data Collection

Sources
- Voluntary submissions from professionals
- Research institutes and bodies
- Research networks
- Financing agencies
- Patient groups
- European Commission

Validation
- Continuous updates
- Validations by professionals
- Scientific consultants in each country
Bimonthly Newsletters

- Key research findings
- New syndromes, genetic discoveries
- Calls for collaboration
- Calls for recruitment
- Scientific meetings and events
Open publication for:
- clinical articles
- research articles
- methodological articles
- review articles
- negative results of clinical trials

Key figures
- 51 articles published in 2006
- 20% highly accessed
- More than 10,000 visitors per month
A Tool for Project Development

Partnership with LEEM and financial support of DG Recherche

Identification of Drugs with Potential for Rare Diseases Previously Authorised for Another Indication

- What are the products?
- Statute of marketing: market authorization, interrupted production
- Clinical experience
- Biographical references

Key figures
131 drugs
with 215 potential rare disease indications
Identification of Clinical Research Barriers

Difficulties in:
- organisation of research
- following of clinical trials
- finding partnerships
- patient recruitment
- development of protocols
- funding for clinical trials
- identification of and collaboration with research centres
- administrative channels

Identification of Products Under Development

- Diagnostic tools
- Monoclonal AC
- Oligonucleotides …
A Database of Innovative Projects

Database

- Research projects with potential indications for rare disease diagnosis or treatment
- Molecules with potential orphan indication
- License offers in France and Europe

Networking

- Establishing relationships between academic research and industry
Users register with OrphanXchange

www.OrphanXchange.org
Innovative Projects

Projects with potential indications for rare disease diagnosis or treatment (129 projects)
- new products
- drugs with alternative indication

Étape 1
Project search

Étape 2
Selection of projects by disease category by product type

Étape 3
Request for contact with researcher or clinician

Researchers/clinicians
Networking

- Seeking contacts, both in and outside Europe
- Putting representatives in contact, organising meetings
- Establishing relationships between academic researchers and industry professionals

- Provides the opportunity to bring together
  - biotechs and investors
  - industry and clinical networks
OrphanXchange figures

Website use

- 215 registered users
- 31 countries: Europe, USA, Japan...
- 46% private sector: industry, biotech, investors
- 500 visits per month
- 113 contacts established

Placement

- Pro-active go-between facilitating partnerships
Thank you for your attention