



## National Clinical Programme for Rare Diseases

### Orphanet: Overview for Clinicians

#### What is Orphanet?

Orphanet ([www.orpha.net](http://www.orpha.net)) is an international rare disease website for clinicians and patients and is the largest repository of rare disease information globally. A total of 39 countries participate in Orphanet including EU countries, Canada and more recently, Australia and Japan. Each participating country hosts an Orphanet team that maintains a directory of resources in their own country. Orphanet is headquartered in Paris, from which it manages the epidemiological data, encyclopedia, classifications and nomenclature, orphan drug designations, as well as information about genes and inheritance.

#### Orphanet provides **direct online access to:**

- ✚ An inventory of rare diseases, with associated genes
- ✚ A support-to-diagnosis tool
- ✚ Clinical and emergency guidelines
- ✚ Specialised clinics
- ✚ Medical laboratories & diagnostic tests
- ✚ Patient organisations
- ✚ An encyclopedia for the general public
- ✚ An inventory of orphan drugs
- ✚ Ongoing research projects including clinical trials
- ✚ Downloadable reports on rare disease prevalence, registries, orphan drugs etc
- ✚ Many links to related information

#### Orphanet Site Stats 2014

- ✚ Average daily site hits = 41,000
- ✚ 2/3 healthcare professionals
- ✚ 1/3 patients and families
- ✚ Enquiries from 226 countries
- ✚ 32.4 million Orphanet pages
- ✚ 11.8 million PDF documents

#### Register Your Expertise on Orphanet

- ✚ **Professionals** can register if they are associated with at least one rare disease activity, whether it be expert centre, laboratory, patient organization, research project, clinical trial, registry or biobank, or as orphan drug sponsorship.

- ✚ Activity is **registered by diagnosis** or diagnoses

#### ✚ **Criteria for inclusion in Orphanet**

- Experts at management and care, at a national or international level
- Multidisciplinary treatment clinics
- Teaching and education
- Collaboration with patient groups
- Research involvement
- National or international collaboration

- Pathways for diagnosis and treatment, including with primary care

✚ **Expert centres** meeting some of these criteria can apply to be registered on Orphanet and after validation by the National Coordinator and Orphanet centrally.

Centres that consider that they may meet all of the European Union Committee for Expertise in Rare Diseases (EUCERD) criteria can also apply to the National Orphanet Team to complete the full EUCERD application form to be named as a National Centre of Expertise. Centres that achieve National Recognition have a symbol next to their Orphanet entry.

✚ **Laboratories:** both research and clinical laboratories may apply to have their rare disease activity listed on Orphanet. Orphanet has replaced EUROAGENTEST as the record of External Quality Assessment and National Laboratory Accreditation, and both INAB and EQA qualifications are shown next to Orphanet lab entries.

✚ **Patient Organisations:** must be registered charities and provide information and support.

The Orphanet Ireland team has started working from the National Rare Disease Office since June 2015 and are beginning to update information on Ireland. Orphanet is a reference for other HSE services such as the Cross Border Directive and the Treatment Abroad National Contact Point.

### **Benefits of registering on Orphanet Ireland**

- ✚ Increased access to and visibility of expertise in rare diseases for patients and healthcare professionals
- ✚ Increased visibility to potential national and international research collaborators and groups, as well as potential rare disease research participants searching for a research project or clinical trial
- ✚ Promotes networking of clinical trials and registration of local or national projects may enhance international 'trial readiness' capacity in Ireland

### **How to register on Orphanet**

- ✚ Register on [www.orpha.net](http://www.orpha.net) **Register my Activity** button
- ✚ It may take some time to complete the online registration forms. The Orphanet team may be able to assist you to upload data, if appropriate.
- ✚ Once registered on Orphanet, you can log in to update the information at any time.
- ✚ Once registered you will only have to respond to an annual email asking you for relevant updates or changes.

The Orphanet team is available to make presentations to faculties, departments or research divisions, or to liaise with you directly about how best to enter expertise information.

Contact the Orphanet Ireland team at [orphanet.ireland@mater.ie](mailto:orphanet.ireland@mater.ie) or 01 809 7475.