

orphanet An Overview for Healthcare Providers

What is Orphanet?

Orphanet is an international rare disease online resource for healthcare providers and patients and is the largest repository of rare disease information globally. Thirty-eight countries participate in Orphanet including EU countries, Canada, Australia and Japan. Each participating country hosts an Orphanet team to collect information about rare disease resources in their own country to be included to the database. Orphanet headquarters in Paris contribute epidemiological encyclopaedia, classifications, and nomenclature, orphan drug designations, as well as information about genes and inheritance. Orphanet Ireland is located at the National Rare Disease Office.

Orphanet provides curated, reliable online information about:

- An inventory of rare diseases, with associated genes
- An encyclopaedia for the general public
- Clinical and emergency guidelines
- Information about specialist clinics
- Information about patient organisations
- Information about medical laboratories and diagnostic tests
- Information about research projects and clinical trials
- Reports on rare disease prevalence, registries, orphan drugs
- Information about members of European Reference Networks

Orphanet Statistics 2018

- 92,740 average daily site hits
- Accessed from 236 countries
- Users 40% healthcare professionals, 35% researchers and industry, 25% patients and families
- 32 million Orphanet pages viewed and 7.5 million PDF documents downloaded

Orphanet's registered Irish expertise (Jan 2020)

- 76 clinics for multidisciplinary management
- 69 rare disease patient organisations
- 25 ongoing research projects
- 100 clinical trials, 34 now recruiting
- 444 professionals registered on Orphanet

What types of information can be registered on Orphanet?

- **Individual healthcare professionals** can register if they are associated with at least one rare disease activity, whether providing clinical management, diagnostic laboratory services, research projects, clinical trials, registries or biobanks, or as an orphan drug sponsor. The rare diseases declared as their expert area are also listed so that the relevance of their Orphanet information is clear to users.
- **Rare diseases clinical services** can register provided they meet the following criteria:
 - The service offers multidisciplinary clinical care for rare diseases
 - Members of the team have recognised expertise in their area and are involved in national or international collaborations
 - There service plans for transition of management from paediatric to adult care
 - The service collaborates with patient groups
 - The service is involved in training or education initiatives
 - The service conducts research

- **Clinical laboratories** can register their diagnostic activity by rare disease. Orphanet has replaced EUROAGENTEST as the record of External Quality Assessment (EQA) and Irish National Laboratory Accreditation (INAB), and both INAB and EQA qualifications are shown in association with the Orphanet laboratory entries.
- **Research and clinical trials** with a rare disease focus, whether basic or clinical, can be registered on Orphanet. The start and end dates, recruitment status, a description of the project and contact details of the investigator and funder are required. This detailed information is helpful to Orphanet users and contributes to the overall picture of the rare disease research landscape in Europe and internationally.

Benefits of registering on Orphanet

- 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

- Further details and registration forms are available on request from the Orphanet Ireland team. Please email orphanet.ireland@mater.ie or call 01 809 7475.
- Once the Orphanet Ireland team approves the applications, the information is uploaded to the Orphanet website.
- Contributors receive an annual email from Orphanet requesting an information update, or entries can be updated at any time via orphanet.ireland@mater.ie

Contact details

To search the Orphanet database www.orpha.net

For Irish rare disease events and news visit our website at www.orpha.net/national/IE-EN

To register your expertise contact the Orphanet Ireland team at orphanet.ireland@mater.ie or 01 809 7475.

The Orphanet Ireland team is available to make presentations to faculties, departments or research divisions, or to liaise with healthcare providers directly about how best to register on Orphanet.

Orphanet Ireland is proudly supported by the National Rare Diseases Office (NRDO). The NRDO is a HSE service, which provides current and reliable information about genetic and rare diseases free of charge to patients, families and health professionals. Find out more at www.rarediseases.ie

