

# The ORPHANET Directory of Services: Activities Related to Rare Diseases Across the UK

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## Background

*Orphanet is an information centre for the rare disease community.*

Orphanet is the non-profit, European Commission funded, official portal offering information and specific services for rare diseases (a rare disease is one which affects less than 1 person in 2000). The website, which is now the largest database dedicated to rare diseases in the world, is accessed daily by more than 20,000 users from 170 countries. The website is freely available in a choice of six languages: English, French, German, Italian, Portuguese and Spanish.

The objectives of Orphanet are to:

- Contribute to improving diagnosis, treatment and management of patients suffering from rare diseases
- Accelerate the development of research
- Reinforce the participation of concerned rare disease stakeholders
- Improve existing resources

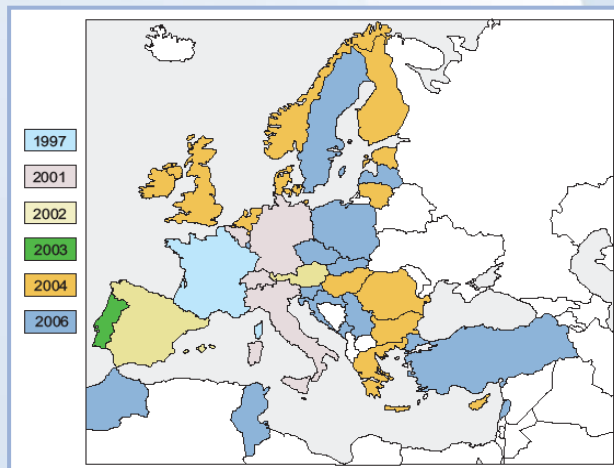
Orphanet offers:

- A list of more than 7000 rare diseases, over 2000 of which feature encyclopaedia entries describing the condition. Entries for the encyclopaedia are produced by international experts in the field and validated by an editorial committee.
- A directory of services providing information on specialised consultations and centres of reference, clinical laboratories, research projects, clinical trials, professional networks, disease registries and patient support organisations.
- Many related services such as the bi-monthly OrphaNews Europe e-newsletter, the peer reviewed academic journal, Orphanet Journal of Rare Diseases and a registration service where patients can volunteer to participate in relevant research projects and clinical trials.

For more information on the Orphanet database or any related services, please visit the website ([www.orpha.net](http://www.orpha.net)) or contact the Orphanet team in your country.

## The Orphanet Consortium

The map below illustrates the 35 Orphanet partner countries and the year in which they joined the consortium. In addition to collecting data for the directory of services in their respective countries, the partners also ensure the relevant professional, patient and public communities in their country are aware of the Orphanet portal and related services.



## Collection of Activities for the Orphanet Directory of Services – Progress in the United Kingdom

The directory of services is a vital part of the service Orphanet provides:

- It aids health professionals and patients by providing information on currently available clinical and laboratory diagnostic services.
- It facilitates collaboration in the research and development of future treatments for rare diseases.
- It allows patients and their families to feel less isolated by providing the means to find relevant support organisations.

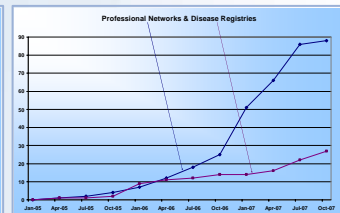
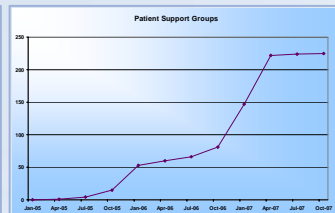
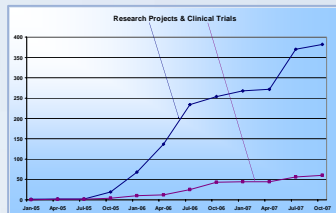
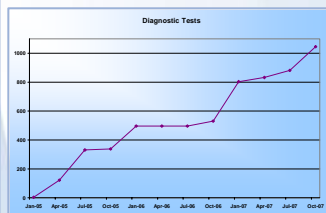
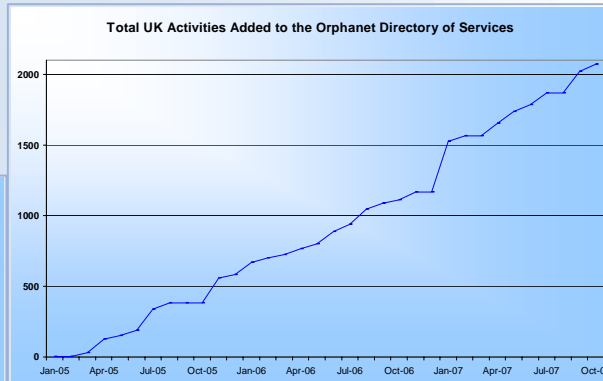
The Orphanet partners are responsible for gathering information for the directory of services from their respective countries (see map). The Orphanet directory of services currently contains the details of over 26,000 activities related to rare diseases.

The UK Orphanet team began collecting the details of any ongoing relevant activities from health professionals and patient support organisations in January 2005. As they were collected, these activities were added to the Orphanet website. The graphs illustrate the progress of data collection in the UK since January 2005 for each activity type.

The collection of UK activities has so far been successful.

Highlights of the activities collected so far include:

- Every test provided by the UK Clinical Molecular Genetics Society network of laboratories is now listed on the Orphanet database.
- Orphanet contains details of the outpatient clinics provided by over 75% of UK Clinical Genetics centres and over 50% of UK National Specialist Commissioning Advisory Group (NSCAG) services.
- Over 90% of UK patient support organisations are now listed in the Orphanet directory of services.



## Conclusions & Future Prospects

This poster presents the data on rare disease-related activities that has been collected to date from across the United Kingdom. This data has been added to the freely available Orphanet website and each activity has been linked to the relevant rare conditions. In total, details of over 2000 activities have been collected from 900 individuals from the UK rare disease community. In addition, the first annual update of data has been completed for the UK. Overall, this represents good coverage of the ongoing UK activities which are related to rare diseases.

In the future, the collection of UK activities for the Orphanet directory of services will prioritise:

- The completion of the diagnostic testing directory including the relevant cytogenetic, biochemistry and haematology tests.
- The completion of the specialised outpatient clinic directory.
- The production of a more comprehensive research project and clinical trial directory.

## How to Register Your Activity:

1. Go to [www.orpha.net](http://www.orpha.net) and click on the English version. Then click the 'Register your activity' button and open the relevant online form. Complete the registration form and click submit.

Register your activity

2. Contact the UK Orphanet team using the details below for a registration form or to register your activity over the telephone.