

EuroGentest/Orphanet Database: new services

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Genetic tests are now offered internationally, through both public and private sector genetic testing services. Physicians prescribing these tests and biologists receiving the samples need to know which tests are available, where they are offered and whether the identified laboratories meet quality standards. To meet this need, Orphanet (www.orpha.net) set up a database of clinical laboratories in the field of rare diseases ten years ago. The data collection covered one country in 1997, fifteen by 2003, twenty-six by 2006 and thirty by 2007. This major effort was made possible by collaboration with EuroGenTest, a network of excellence in the field of genetic testing, supported by the European Commission.

Currently, the database includes data from 1,233 laboratories offering 16,336 tests for 1,504 diseases. The collection of data occurs at the national level through partnerships with key leaders in the field. Laboratories are requested to fill in an extensive questionnaire (either online or by traditional means). The response rate is approximately 80% after three reminders. All the information is updated yearly through an online questionnaire already containing the existing information, in order to minimize the workload for the laboratory.

In March 2008, Orphanet launched a new version of its website to improve its user-friendliness and to better served its end-users.

The new services include the possibility of query by gene (symbol or name in English) in addition to the traditional query by disease name. It also includes the possibility of query by broad category of diseases as classifications have been introduced in the database.

The data on laboratories have been expanded to provide information on quality management schemes that the laboratories have put in place.

This information is widely used as Orphanet is accessed daily by over 20,000 users, of which 20% are seeking information on genetic testing.

These new services are expected to promote the quality laboratories.