

European Directory of DNA diagnostic Laboratories (EDDNAL)
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Executive summary of the Final Report

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**Programme of Community Action on Rare Diseases
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Summary

This final report presents an account of the activities of the EDDNAL project for the year 2001-2002. Details are given concerning the development of the project as well as an assessment of usage of the EDDNAL web site as a proxy for impact.

EDDNAL is a non-profit registry, supported by grant from the EC, which is specifically intended for professional use. The information contained in EDDNAL is published as an online clinical resource (www.eddnal.com). It provides standardized information on diagnostic services for heritable syndromes and disorders offered by laboratories throughout fourteen EU countries as well as Switzerland, Norway, Poland. At present, the directory contains 337 laboratories, 906 genetic conditions and 756 contact persons.

An update, begun in 2000, has enabled the registry to collect more information on the diagnostic procedures performed by each laboratory, quality assessment scheme participation and logistical details concerning the organization of testing. The update has also acquired information on research being carried out within each laboratory.

In the past, the EDDNAL web site lacked certain functional parameters that would have enabled the publication of this information. Because the new updated laboratory information introduced more subtleties and details than the past web site could accommodate, a more sophisticated web application was required in order for the medical community to have access to this information. The EDDNAL administration negotiated a contract with an informatics company and the new site was officially launched in October 2002. The feedback from the scientific community has been entirely positive.

Throughout the past two years, the EDDNAL resource persons have been engaged in multiple other endeavours. The literature on gene discoveries and research is constantly reviewed with the aim of identifying and approaching laboratories which may provide diagnostic testing for inherited disorders but are as yet not included in the registry. In the past two years over 200 new laboratories have been invited to apply to be included in EDDNAL. The overall response has been positive. The EDDNAL Registry Manager has given multiple oral and poster presentations in diverse venues spanning both the European as well as North American continents.

Statistics on the use of the web site indicate a growing interest in and use of the information provided by the web site. In addition, these statistics show that the audience using the information in the EDDNAL registry, while primarily European, also come from North America, Australia, Oceania and Asia.

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