**Executive summary of the**

# FINAL TECHNICAL REPORT

# PROJECT : Network of Public Health Institutions on Rare Diseases (NEPHIRD)

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**Project leader:** **Domenica Taruscio**

**Institution:** **Istituto Superiore di Sanità**
**Viale Regina Elena, 299**
**00161 Roma – Italy**
**Tel. +39 06 49902805**
**Fax +39 06 49387140**
**e-mail: taruscio@iss.it**

## SUMMARY

Network of Public Health Institutions on Rare Diseases (NEPHIRD) is one of the nine projects financed by the European Commission following the decision (No 1295/99) taken by the European Parliament and Council to launch a programme of community action on rare diseases (RD), within the framework of action in the field of public health. NEPHIRD has the objective of developing models to define the epidemiological parameters of RD. Besides, the project aims at describing the situation, in terms of public health initiatives, of RD in the participating Countries as well as at developing an interactive web-site dedicated for exchange of experience and diffusion of information.

Public health institutions from 15 European Countries participated in the project which is coordinated by the Istituto Superiore di Sanità.

A web-site dedicated to the project is already developed and put in place (http://www.cnmr.iss.it/NEPHIRD/index.htm). Two questionnaires are administered – the first on various aspects of RD's problem in the participating Countries and the second as an inventory of diagnosing centres and possible sources of systematically collected epidemiological data on eight RD that were identified to represent different epidemiological realities. Following the meetings of the project management group, a general meeting (Steering committee) was held where all project participants were invited to share the results and their experiences.

The results of the questionnaire N. 1 indicated that public health initiatives have been taken recently in few European Countries, though such initiatives are not homogeneous. The inventory showed that several centres exist which handle a significant number of patients and collect epidemiological data based on local initiatives. The general meeting addressed the issues of definition and classification of RD, the possible constraints for data collection and the quality of data.

The Expert Meeting on Sociological Issues discussed such items as: who is involved in supporting people with RD, the need for a new language and more positive and accurate terminology; the need to reflect the experiences of people with RD (e.g., through expoiting the use of narratives); and finally, some key reflections to develop a culture for RD.

As concerns the role of patient's registries in the epidemiological data collection on RD, the activity of *UK Society far Mucopolysaccharide and Related Diseases Patient Registry* has been evaluated as a positive model, able to promote and support research.

Data collection on RD is relatively difficult from both the technical and the resource standpoints; efforts should be made to promote quality assurance and implement usefulness both in terms of public health and epidemiology. Overall, the consensus position was that the approach in the field of RD has to capitalise local efforts dispersed with in the European community and associated Countries.