The Italian National Centre of Rare Diseases

**Activities of the Centre**

National and international activities on rare diseases performed by the Italian National Centre of Rare Diseases are described. The main activities of the Centre are:

- Surveillance of rare diseases at national level (Italian National Registry Rare Diseases);
- Prevention activities;
- Training and continuous education for health workers;
- Information for patients, relatives, and population;
- Elaboration and diffusion of clinical guidelines;
- Accessibility and quality to health social services for the patients with rare diseases;
- Quality assurance and standardization of genetic tests;
- European project Network of Public Health Institutions on Rare Diseases (NEPHIRD).

**The National Registry for Rare Diseases**

The National Network for Rare Diseases includes regional centres (for prevention, diagnosis, treatment of rare diseases) and the National Registry. The aims of the Network are:

- To improve prevention activities, to guarantee an appropriate and timely diagnosis and treatment and to realize the surveillance of rare diseases.
- To elaborate diagnostic protocols and clinical guidelines.
- To estimate incidence or/and prevalence.
- To improve collaboration among health care operators to find consensus on diagnostic criteria.

**Primary Prevention of congenital defect and folic acid**

The Italian National Centre for Folic Acid Promotion started in April 2004, in order to integrate and optimize activities present at local or regional level. The Network involves participants from research institutes, the Health Ministry, Regions, local health services, Universities, as well as physicians, journalists and representatives of patients associations. The Network has already elaborated and diffused a National recommendation to promote an increased intake of folic acid among women in fertile age.

**Training and continuous education for health workers**

Numerous Courses, meetings and workshops are organized from National Centre of Rare Diseases, to provide information to health professionals, involved in epidemiological and clinical activities.

**Information for patients, relatives, population**

The National Centre of Rare Diseases provide information and support to patients, families and population. Moreover, it collaborates with Patients associations to spread knowledge on rare diseases through web site [http://www.cnmr.iss.it](http://www.cnmr.iss.it) in which a specific section is dedicated to National associations of patients.

**Guidelines**

The National Centre of Rare Diseases is engaged in elaboration and diffusion of multidisciplinary guidelines for assistance of persons with Down syndrome and their relatives.

**Quality Assurance and Standardization of Genetic Tests**

The Italian External Quality Assessment (I-EQA) started as a research projects, financially supported by the Italian Ministry of Health (“Italian national project for standardisation and quality assurance of genetic tests” (D.lg 505/92) and “Genetic tests: from the research to clinic”) and coordinated by the Istituto Superiore di Sanità (ISS). Four trials have been performed and completed (I-EQA-1: 2001; I-EQA-2: 2002; I-EQA 3: 2003 and I –EQA 4).

**NEPHIRD**

NEPHIRD is a network of Public Health Institutions working on rare diseases (RD) in Europe funded by the European Commission (DG SANCO, 2002-5). As the name implies NEPHIRD was conceived as a forum for public health institutions where sharing of opinions and experience will be place. Public Health Institutions from 15 European Countries participated in the project which is coordinated by the Istituto Superiore di Sanità. The main aims of NEPHIRD are: epidemiological data collection and assessment of public health aspects (access to health and social services, quality of life, etc.) of RD.