Domenica Taruscio, Salerno Paolo, Kodra Yllka, Agazio Elvira
National Centre Rare Diseases, Istituto Superiore di Sanità – Rome, Italy

Introduction
The Italian Government promulgated a Regulation on Rare Diseases (Ministerial Decree 279/2001, «Institution of the national network of rare diseases»). The National Network includes regional centres and the National Registry of Rare Diseases. 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.

The Italian Government underlined equity in the availability of health services as a core element of the 1998-2000 and 2003-2005 National Health Plans. Accordingly, the Government identified rare diseases as a priority area and emanated regulations referring to these conditions. The main element identified and endorsed by the Health Ministry is the institution of the National Network for Rare Diseases and the National Registry of Rare Diseases established at the Istituto Superiore di Sanità-Rome (Minister Decree, 279/2001 «Institution of the national network for rare diseases») (Table 1, 2, and 3).

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.

The Registry has been established at the National Centre Rare Diseases at the Istituto Superiore di Sanità for collection and elaboration of epidemiological data. The specific objectives of the Registry are:
- To estimate incidence or/and prevalence
- To elaborate diagnostic protocols and clinical guidelines
- To improve collaboration among health care operators to find consensus on diagnostic criteria.

Results
The Registry is collecting epidemiological data at National level on 547 different rare diseases. Analysis have been performed for specific diseases on collected data to elaborate ad hoc studies, regarding for example:
- diagnostic delay: in Prader Willi syndrome the delay has been calculated as about 14 months;
- patients’ migration: we analysed 207 patients affected by rare metabolic diseases and we found that approximately 40% of them migrate through Italian Regions to obtain the diagnosis.
- Collaboration with the Rare Diseases Registry of the Marche Region (Table 4, 5, 6 and 7)

Conclusions:
Epidemiological tools, such as Registries, are very useful to plan public health activities at regional, national and