Patient care from rare disease to health management: networks of care and research

G. Annicchiarico

Regional Coordination of Rare Diseases, Regional Health Agency Puglia, Italy

In Italy the Ministerial Decree n. 279/2001 regulated for the first time the assistance of patients suffering from rare diseases. Rare Diseases are one of the main objectives of the last National Health Plans. For their intrinsic characteristics, these diseases have in common only epidemiological data. Rare Diseases (RD), according to a definition adopted within the European Community, have a population prevalence of less than 5 cases per 10,000 individuals. The international community does not share a unique opinion. These are heterogeneous pathologies, joined by similar welfare issues, which need to be addressed globally and which require a particular and specific care, for their diagnostic difficulties, clinical severity, chronic course, disabling outcomes and burden of treatment. In Europe it is estimated that the number of people suffering from rare diseases fluctuates between 27 and 36 million. But there is a paradox: there are a lot of rare diseases, but there are few or very few patients for each disease. Europe recommended to individual States a comprehensive approach for the definition, by the end of 2013, of the National Plans that integrate present and future strategies, at local, regional and national level. In Italy, a State-Regions agreement in 2007, marked the birth of the Regional Coordination Centre of Rare Diseases and the Regions, responsible of patients care, have identified in their territories Centers of Experience, health centers dedicated to rare diseases or groups of diseases. The structural characteristics of the Italian Public Health System and the assistance requirements of children and people affected by rare diseases, prompted the establishment of integrated welfare systems within the networks and between networks: the hospital network, the local primary care network, the hospital and local pharmacies network, the palliative care network, municipalities, Disability – Handicap care networks. Actually, it is now possible in the regions with the longest experience, with the support of safe logical infrastructure, to survey online patients needs and to direct the programmatic decisions of regional governments. At present seven Italian regions use, with the same information system, the same working platform, that, once updated, will allow 23 million people to share the same rights and the same treatment protocols. To date Italy is the repository of an innovative model of care that connects the vertical/hospital system of care with the horizontal/local one. The regions regularly compile the National Registry which is currently in charge of the Italian Institute of Health. In many cases, the geographical dispersion of patients is one of the major limits to recruitment in clinical trials, and to support in basic, clinical or translational research. Therefore, appropriate networks of care are required, besides Italy, between States and even between continents (1). In 1997 Orphanet was born in France. In 2011, the project was expanded to 37 countries of the EU (Journal of the European Union 23/02/2009) – Joint Action. In 2007, the Executive Agency for Public Health of the European Commission funded for three years Dyscerne, a network of centers of dysmorphology. More than ten years after the first European recommendations to orient the decisions of individual States, it is time for new agreements which will establish novel networks of care and research.

(1) The case for a global rare-diseases registry
Forrest et al., Lancet 2011, 377: 1057-9.