The Italian National Plan for Rare Diseases

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Introduction

Italy was one of the first Member States (MS) in the European Union (EU) to regulate the field of rare diseases (RD). Since 2001, many initiatives were carried out at central and regional level. The development of a national plan or strategy for RD stems from the necessity to fulfill the EU Commission Recommendation to MS to adopt national plans or strategies for RD by the end of 2013[1]. However, the adoption of a systematic measure, logical and coherent with planned strategies and actions at all levels, was demanded by national stakeholders in the field of RD.

The first version of the document was drafted by the General Directorate of Planning at the Ministry of Health (MoH) in collaboration with small groups of experts. It was submitted to all the stakeholders (patient and family organisations, healthcare professionals and professional bodies) during a brief consultation process. The document considered also previous requests from the National Board of Patients with RD (http://www.cndmr-insieme.it/) and patient organisations (Federation of the Organisations of the Italian Patients with RDs - UNIAMO and Italian Movement of Rare Patients - MIR). These requests had been previously presented at the final national conference of the EUROPLAN project in 2012[2].

The Italian legal framework on rare diseases

Coming into force just before the approval of the Regulation (EC) n. 141/2000 of the European Parliament and the Council on orphan medicinal products[3], the Italian legislation in the field of RD derives from the willingness to combine the organisation of healthcare assistance with all aspects related to the rights of citizens affected by RD. Specifically, the Legislative Decree n. 124/1998[4] introduced the new discipline on exemption from co-payment for health services nationwide and led to the adoption of the Ministerial Decree (MD) n. 279/2001[5], which is the first regulatory act in the field of RD. The decree identified a list of life-threatening or chronically debilitating diseases with both a high level of complexity of care and a prevalence of not more than 5 in 10,000 inhabitants, in accordance with the EU definition. The diseases included in the above-mentioned list are formally called as "rare" and totally exempted from the co-payment for outpatients benefits provided by the National Health Service (NHS) for diagnosis and treatment. When necessary for patient's diagnosis, also genetic testing to blood relatives is delivered without any charge. Medicinal products are not the subject of the decree and are provided in accordance with Italian Medicines Agency (*Agenzia Italiana del Farmaco*, AIFA) classification. Moreover, the MD 279/2001 establishes a specific network for RD consisting in both accredited health facilities identified by Regions, which are responsible for the diagnosis and treatment of RD and the systematic collection of data concerning RD resulting in the National Registry of Rare Diseases (NRRD) at the Italian National Institute of Health (*Istituto Superiore di Sanità*, ISS)[6].

The effectiveness of MD 279/2001 was confirmed after the approval of the Constitutional Law n. 3/2001 that conferred to the Regions the entire responsibility for the organisation of the healthcare assistance, the planning and management of their own healthcare activities, as well as the provision of healthcare services to the population through Local Health Units. The central State (namely the MoH) has the responsibility to define the general principles and priorities and to establish the national healthcare entitlements (Essential/Basic Levels of Healthcare Assistance, LEA), which are financed by general taxation. Moreover, the MoH is required to ensure that LEA are provided to all citizens and that the Regions have the appropriate financial resources. Finally, the main guiding principles of the NHS are those of universality, equity and solidarity and the LEA provided to people with RD are the same, but free, national healthcare entitlements which are guaranteed to all citizens according to criteria of effectiveness, appropriateness and quality, as well as quantitative and qualitative standards and appropriate way of delivery. On the other side, Regions are allowed to provide further services, as far as they can cover the costs with their own economic means. In recent years, many Regions decided to supply "extra-LEA services" to their population. Among the "extra-LEA services", some Regions include free access to specific further benefits for people suffering from a RD, such as some medicinal products currently delivered with fee but also healthcare assistance to further diseases not currently included in the national list of RD.

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The contents of the draft of the National Plan for Rare Diseases

The first part of the National Plan for Rare Diseases (NPRD) recalls the European and national legislation and a description of the main areas of intervention: LEA; the organisation of the system and its components (the National network for RD, the NRRD, the Congenital Malformations Registers (CMR), and Disease-specific registers, the National Board of Regional Health Authorities on RD, biobanks, coding and classification of RDs); the healthcare and pharmaceutical pathways for RD patients; the cross border healthcare; the role of the patient alliances; research; training and education of professionals, and information; prevention activities.

It is important to highlight that any social intervention is not included in the current NPRD.

The second part of the NPRD "Objectives and monitoring" links each of the above areas of interventions with specific actions for the 3-year period.

The network organisation, set up with the MD 279/2001, deserves special consideration.

Despite the exclusive competence of the Regions established by the aforementioned Constitutional Law, a supra-regional level is recommended for very RD ensuring follow-up activities to be performed near the patient residence. An inter-regional organisation may allow different but appropriate treatment of patient needs according to the level of know-how of each health professional.

The model aims at guaranteeing both high specialised functions and assistance to general needs, available at the healthcare district level.

Identified units (preferably hospital units) are the national network hubs, specifically detected among those who can provide documentary evidence of expertise in diagnosis and care of a specific rare disease or groups of RD, as well as suitable equipment and complementary services to assist in emergencies and to perform molecular, genetic and biochemical diagnoses. These facilities can be guaranteed also by means of functional relations with external services, such as genetics centres and become integral part of the RD network. Furthermore, the identified units are required to comply with the clinical protocols set up in accordance with the clinical guidelines issued by the scientific societies. In addition, all units must also cooperate with general practitioners and local services to provide personal, comprehensive and continuing care to patients. Finally, the national network must fulfill the EU requirements in order to comply with the Council Recommendation on an action in the field of rare diseases (2009/C 151/02)[7] adopted on June 8th, 2009 along with both the EUCERD Recommendation[8] concerning the quality criteria for Centers of Expertise (CEs) in Member States and the implementation of European Reference Networks (ERNs)[9]. All its identified units, firmly included within the regional networks of care, must be able to perform the duties expected from European CEs.

In the assessment process of these units, Regions can use multi-stakeholder opinions, including patient organizations; benchmarks and indicators should cover processes, outcomes and impacts, such as patient reported outcomes. An appropriate governance is ensured through the establishment of a National committee, involving the competent Ministries, the Regions and AIFA, as well as the National Agency for Regional Health Services (*Agenzia Nazionale per i Servizi sanitari regionali*, Age.Na.S.) and patient alliances. The Committee has the task to both define the main strategies for diagnosis and care, research and social promotion, adapting them to the specific economic resources and to optimise the most appropriate monitoring actions.

All services and national healthcare entitlements, including those regarding RD, are currently funded by general taxation and the NHS financial budget is yearly distributed by the central Government to the Regions. The specific expenditure for RDs is difficult to quantify. However, since medicinal products are among the major invoices of expenditure, an accurate medicinal products' accountability may help to predict a great part of the real expense for the NHS.

In recent years, the situation has become more complex and more heterogeneous among Italian Regions, due to the dramatic differences of Regional Health Services budgets. In order to address this scenario, the Central Government has enforced a series of so-called Budgetary Balance Plans aimed at solving the problems of insufficient financial coverage for services provided in some "very critical Regions". The Plans introduced substantial budgetary limits and measures to contain their healthcare cost levels and trends. As a consequence, many Regions undergoing a Budgetary Balance Plan have been forced to cancel the extra-LEAs services they were previously providing, including services for people suffering from RD.

Conclusions

The MoH is aware that expectations of the main stakeholders, in particular of patient associations, might be excessive compared to the actions proposed in the NPRD draft, mainly because of the limitation of financial resources.

The possibility to further allocate dedicated funds for RD depends on the overall evaluation of the central Government. Indeed, the draft of the NPRD suggests to include RD among the performances that are being verified by the national committee for the evaluation of the provision of LEA.

However, the draft of the NPRD specifically considers the efficiency and the effectiveness of the general organisation and the national network to minimize the consequences of budgetary limits. Moreover, it emphasises the importance of a strict cooperation and exchange of experiences and best practices among all the identified regional expert units coordinated at a supra-regional level.

Keywords: Essential Levels of Assistance (LEA), extra LEA services, national network, budgetary balance plans.

The Author declares no conflicts of interest.

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