

Set of Rules in Italy for Rare Diseases Management Sanitary Policies and Epidemiologic Data in Puglia

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Abstract:- Rare disease definition takes into account the low prevalence amongst population of each one of these pathologic conditions, despite of high impact of them worldwide. This work starts from detection of many difficulties in rare disease patients affected management and also analyzes progress on rare disease in Italy and in Puglia. Epidemiologic data, collected by new regional informative sanitary system, highlight main problems in diagnostic-care pathways but also assure efficiency of improvement policies put in place from 2001 to nowadays, by definition of Italian and regional sets of rules.

Keywords:- Rare Disease; Direct Distribution; Territorial Pharmacy; Diagnosis-Care Pathway.

I. INTRODUCTION

Rare diseases are a wide and heterogeneous group of pathologic conditions of low prevalence amongst population. In EU definition of rare disease is when it occurs 5 patients on 10.000 inhabitants (0,05%)[1]. World Health Organization (WHO) data report about 8.000 different rare diseases. This is a growing number because of advances in science and genetic research[2]. Worldwide there are 300 mln of people affected by rare disease[3]. More than 30 mln interest UE and 2 mln Italy, according to Orphanet. For rare diseases identification it is used an alphanumeric system: the international classification of diseases (ICD), from WHO. In Italy the ICD 9th revision - clinical modification (ICD-9-CM) consists of 17 chapters, 16 for diseases referring on anatomical site and clinical signs, and 1 for traumatism and poisoning. In Italy the attention for rare diseases grows up in '90 and spreads in latest years, with the awareness that them need specific targeted policies. First limit is difficulty of early correct diagnosis because of complex clinical signs. Diagnostic-care pathways are complicated by lack of reference aids, placed uneven across the territory. Patients and their families live a loneliness experience doubly painful for disease and its rarity, for which it is often unknown and not recognized. For these reasons appropriate information is very important to patients and people around

them. The aim of our work is to offer purposes for system's efficiency optimization starting from the analysis of strengths, weaknesses, opportunity and threats of Italian set of rules. Focus on regional organization has been necessary to understand real problems in rare diseases management.. We analyzed the data of Puglia, of a province such as Bari, and of a socio-sanitary district of the province of Bari which DSS n. 4

II. ITALIAN REGULATION

A. Essential Assistance Levels

National health service (SSN) guarantees to patients sanitary services provided by essential assistance levels (LEA). LEA are defined in decree of council ministers president (DPCM) 29 November 2001, updated by DPCM 12 January 2017. List of rare diseases is updated with more than 100 new diseases. About pharmaceutical assistance, according to current law, class A drugs are provided by SSN, class C drugs are defrayable to citizens. However, due to sanitary regionalization, each region could guarantee further LEA to citizens, if economic and financial conditions allows it.

B. National net for rare diseases

Health minister instituted "national net for rare diseases" with ministry's decree (DM) 279, 18 May 2001 (Fig. 1). It consists of national net's aids (PRN) for prevention, surveillance, diagnosis and treatment of single disease, endowed of support structures, emergency services and diagnostic instrument for biochemical and genetic-molecular diagnosis, distributed on national territory. List of PRN is approved with regional council resolution (DGR) n. 1491 of 3 October 2017 and is updated with DGR n. 329 of 13 March 2018. This organization aims to offer qualified structures for patients care and to increase their territorial distribution for a simple access to aid everywhere in the nation. For this aim, the state-regions and autonomous provinces deal of 10 May 2007 instituted regional and inter-regional coordination centers for PRN link and correct address of patients, according to structure availability and patient/doctor needs.

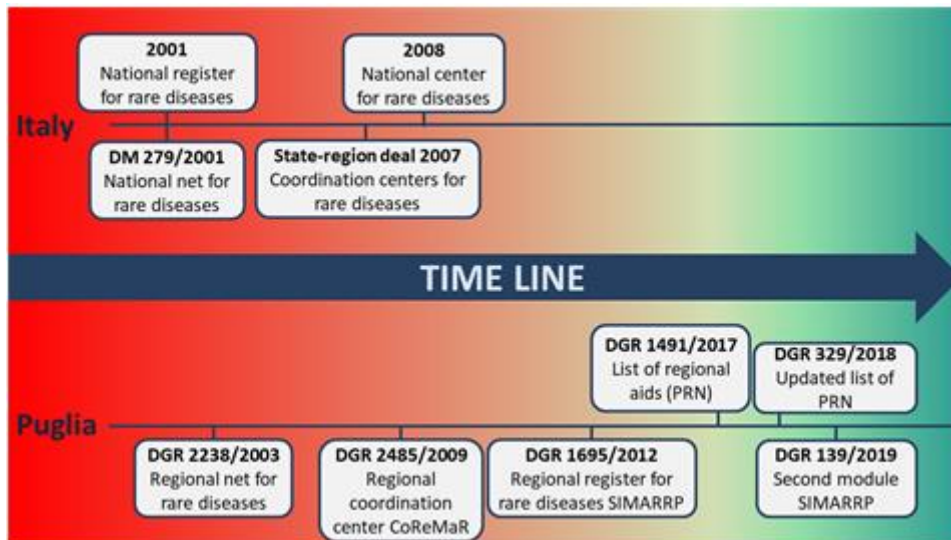


Fig 1:- time line of policies for rare diseases management in Italy and in Puglia.

C. Regional net for rare diseases

In Puglia it has been instituted with DGR n. 2485 of 15 December 2009 updated with DGR n. 2048 of 13 December 2016, regional center for rare diseases coordination (CoReMaR). DGR n. 225 of 23 February 2017 established the creation, in every ASL, of a territorial center for rare diseases (CTMR), for citizens information and patients address to qualified structures for diagnosis and treatment. CoReMaR manages regional register for rare diseases.

D. Focus on ASL Bari

According to the regional procedure, a PTP is drawn up for patients with rare diseases who belong to an authorized regional center. On the basis of this PTP, the services covered by the LEA are guaranteed. About extra-LEA, because of financial condition of Puglia, region is unable to provide them. However, according to ministry of health prescriptions, it is instituted a company commission for evaluation of every PTP, in order to accord extra-LEA dispensation in case of declaration of irreplaceable and indispensability, and clinical documentation of therapy.

The authorized drugs are supplied through the pharmaceutical services of the asl of residence of the patient in direct distribution (DD). In rare diseases monitoring by DD is important. For rare these patients care it is necessary to use a lot of drugs: orphan drugs, off-label therapies, drugs approved by law n. 648/1996, drugs approved abroad, experimental drugs according to decree of ministry of health of 8 may 2003, galenic preparations, and also medical devices, dietary products and other products not classified as drugs. Monitoring system in DD is supported by on-line prescription and dispensation.

III. EPIDEMIOLOGIC DATA

Thanks to new regional sanitary informative system (NSISR) EDOTTO, epidemiologic data on rare diseases in ASL Bari have been collected (Tab I). There are 9791 patients in ASL Bari. ASL Bari consists of 12 DSS. In DSS n. 4 there are 1115 patients. This corresponds respectively to 0,77% and 0,78% of population (Fig. 2). Only 102 of them are in treatment with drugs that withdraw from territorial pharmacy of DSS n. 4; 21 provided by on-line PTP (Fig. 3).

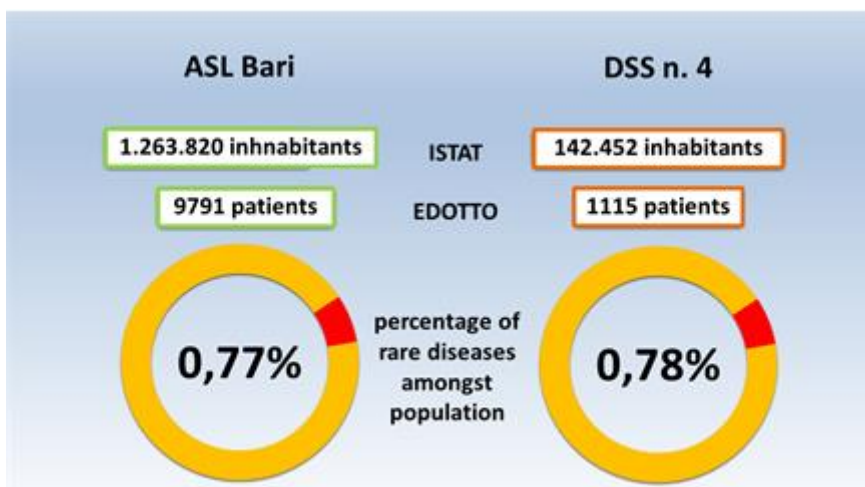


Fig 2:- Percentage of patients with rare disease amongst population. Comparison between ASL Bari and DSS n. 4 ASL Bari. Data patients from EDOTTO; data inhabitants from ISTAT.

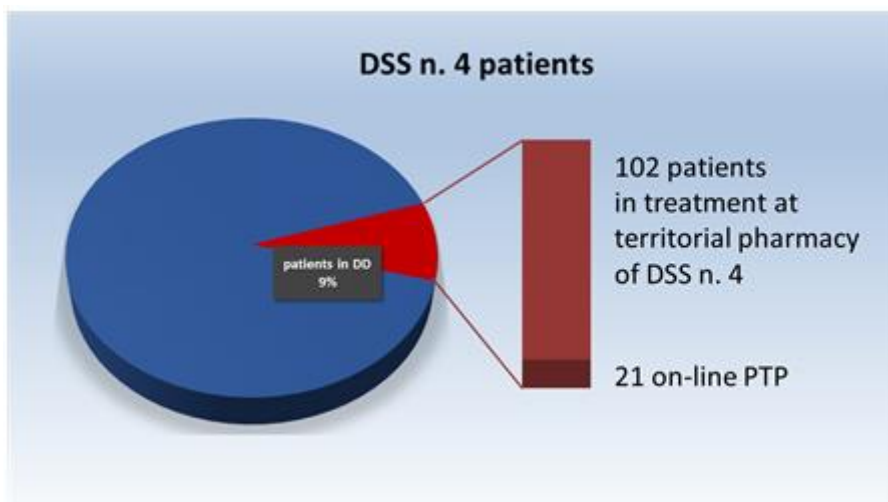


Fig 3:- Fraction of Patients with rare disease in DSS n. 4 ASL Bari in treatment at territorial pharmacy service in direct distribution, after paper prescription or, in a few cases, on-line PTP.

IV. CONCLUSION

When clinicians stare in front of a rare disease, first step is to find the greatest way of treatment, taking information from reliable sources, not always available. Second step is to address patient to adequate aid, regarding territory supply and patient needs. Regional net creation has been very important to overcome these problems, but its empowerment is required, by organization of specialized structures spread all over the nation and linked to each other. In fact principle on which net institution is based on, is that nobody could do everything but cooperation in important. Lack of information is another weakness of the system. In fact from epidemiologic data it results that only 9,15 % of DSS n. 4 patients, are in charge at territorial

pharmacy. The aim of our work is to make information about rare diseases management clear and available for a wider group of persons, clinicians but also patients and relatives or caregivers. Moreover informative systems represent a great resource. The use of these new technologies allows to create diagnostic-care pathways valid all over the nation, shared worldwide and this is warranty of efficacy and safety of the treatment. Use of on-line PTP is important also to guarantee a faster patient taking charge from service for health care supply. However, nowadays in the examined DSS prescriptions are made by on-line PTP, instead of paper form, so operations for clinicians awareness are important. Wish for future is improvement of link aids organization, correct information spreads and informative systems use.

ASL BARI	DSS n. 4 ASL BARI		
EDOTTO	EDOTTO	Direct Distribution	On-line PTP
9791	1115	102	21

Table 1:- Patients with Rare Disease

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