

RARE DISEASES AND CLINICAL RESEARCH: THE 10-YEARS EXPERIENCE OF ASL LECCE ETHICS COMMITTEE

Maria Rita De Giorgio¹, Cinzia Morciano¹, Stefania Di Noia¹, Diana Conte²

¹Segreteria Scientifica del Comitato Etico ASL Lecce, Lecce - Italy, ²Comitato Etico ASL Lecce, Lecce - Italy

Introduction: Rare diseases (RD) include 5000 to 8000 life-threatening or chronically debilitating diseases, globally affecting 27-36million people in Europe. However, clinical research in the field suffers from limited investments and organizational challenges, especially in peripheral areas. In Italy, an inter-regional network has been established since 2001for sharing data and expertise, and a National Registry has been activated. In Puglia, where estimated RD patients are more than 15000, a population registry has been developed, representing a valuable resource for epidemiological analyses and health planning. The Ethical Committee (EC) for the area of Lecce, in the south of Puglia, has over the years recognized a distinctive value to research in RD. The present work aims at describing, from the peculiar point of view of a South-Italian EC 10-years experience, the frequency and main features of clinical studies accessible for RD patients in the area.

Methods: Starting from the EC database of all studies evaluated/authorized between 2008and 2018in the area of Lecce, this report analysed the studies focusing on RD or rare tumours (RT). Four sources have been used for reference: ORPHANET, RARECARE, Health Ministry database of RD, Rete Tumori Rari. The following definitions have been used: RD, defined by a prevalence $\leq 5/100000$ inhabitants; RT, defined by an incidence $< 6/100000$. RD and RT have been analysed separately, describing the frequency and the following features: 1) pathology group; 2) nature and type of promoter; 3) study design.

Results: Between 2008and 2018, the EC for ASL Lecce authorized a total of 830 studies, of which 20 (2,4%) focused on RD and 26(3,1%) on RT. Most studies on RD concerned haematological diseases (70%), more frequently haemophilia, beta-thalassemia, and myelofibrosis, followed by algodystrophy, and amyotrophic lateral sclerosis. These studies had been mainly proposed by non-profit promoters (70%), especially academic institutions (45%), and 80% presented an observational prospective or retrospective design (mostly patient registries), investigating the real-world effectiveness and safety of therapies. As for RT, 84,6% studies focused on haematological/lymphoid tissue neoplasms, primarily myelodysplastic syndromes and mantle cell lymphoma, followed by neuroendocrine tumours. These studies were frequently promoted by non-profit organizations (69,2%), mainly patient associations (57,7%). Compared to RD, 61,5% studies on RT were clinical trials investigating the efficacy of new drugs or combination therapies.

Discussion & conclusion: Among Italian regions, Puglia has effectively developed RD-dedicated policies and networks, with several centres offering assistance and access to research. The main aim of this report was to focus on clinical research accessible for RD patients in the area of Lecce, through the peculiar point of view of an EC. These analyses show that RD-research has been mainly fuelled by non-profit promoters, such as academic institutions and patient associations, which often collaborate with the common aim of testing new therapeutic strategies, especially for haematologic diseases. Most studies had indeed a therapeutic or prognostic aim, while research about diagnostic approaches or health assistance has been limited (globally, 6out of 46studies), and the observational design represented the most frequently chosen option. The investments of private companies have been rising over the years, mostly through funding projects that sustain associations or public research centres. Creating a RD-focusing network among research centres and related ECs in Puglia may help to develop effective strategies for encouraging research in the area and improving access for patients with RD or RT to national and international studies.