



# HEALTH SYSTEMS SUSTAINABILITY FOR RARE DISEASES

Edited by Rita Maria Ferrelli, Bernardino Fantini and Domenica Taruscio

## Preface

Rita Maria Ferrelli<sup>1</sup>, Bernardino Fantini<sup>2</sup> and Domenica Taruscio<sup>3</sup>

<sup>1</sup>*Servizio Formazione, Istituto Superiore di Sanità, Rome, Italy*

<sup>2</sup>*Faculté de Médecine, Université de Genève, Genève, Switzerland*

<sup>3</sup>*Centro Nazionale Malattie Rare, Istituto Superiore di Sanità, Rome, Italy*

A disease or disorder is defined as rare in Europe when it affects less than 1 person in 2000. There are more than 6000 rare diseases. 80% of rare diseases are of genetic origin, whereas 20% are multifactorial and are often chronic and life-threatening. In spite of the limited number of people that can be affected by a single rare disease, the total number of patients living with a rare disease is high: on the whole, rare diseases may affect 30 million European Union citizens. This epidemiologic pattern deserves to be tackled at the European level. The European Commission has been financing several initiatives and Joint Actions with the purpose of providing a better support at European, national and local levels. Among them, the Joint Action RD-ACTION ([www.rd-action.eu](http://www.rd-action.eu)) aimed at ensuring an integrated European approach to the challenges faced by the Rare Disease (RD) community and at promoting, catalysing and triggering multi-stakeholder debates which are necessary both at European level, to build shared strategies, and at national level, to support the integration of EU policies on rare diseases across Member States.

RD-ACTION was co-funded by the EU Health Programme 2014-2020. It started in 2015 and ended in June 2018. RD-ACTION brought together 64 partners – academia, Ministries of Health, health institutes, medical universities and patients' organisations – from 40 countries, including all 28 EU Member States. The strength of RD-ACTION lies in its multi-stakeholder integrated approach and its pan-European and well structured coverage that included, but that was not limited to the European patient organisation for rare diseases (EURORDIS – Rare Diseases Europe).

Among the activities carried out by the Joint Action, the National Centre for Rare Diseases (Centro Nazionale Malattie Rare, CNMR) at the Italian National Institute of Health (Istituto Superiore di Sanità, ISS) was entitled to develop a specific task to identify mechanisms influencing sustainability, equity and resilience of health systems for rare diseases. This monograph aims at collecting issues debated during the progress of CNMR task. All of them represent crucial factors

raised by the study of rare diseases in the light of improving the organization and sustainability of services for patients living with rare diseases.

*Fantini and Vaccaro* review the literature available on value based health care and relate it to RD. Starting from the critics of an purely economic definition of value and healthcare evaluation, efficacy and efficiency, they include the equity dimension in the definition of value-based healthcare and underline the need of radically re-examining how to organise the delivery of prevention, and healthcare services, in the framework of a new culture of health and well-being, to empower and give voice to vulnerable groups.

*Taruscio and coll.* focus on primary prevention as an essential factor ensuring sustainability of health systems for rare diseases. They emphasize an integrated "One Health" approach, linking knowledge and action, as a requirement to get effective primary prevention. The role of surveillance of health events and potential health-damaging factors, science-based risk analysis, citizens' empowerment and education of health professionals is crucial in primary prevention.

*Severin and coll.* stress the role of education in building sustainable health system and allowing better health and well-being for people with rare diseases. They sustain that by giving knowledge, training competences and promoting active participation and responsibility through education, a new dialogue between policymakers, stakeholders, professionals and rare disease patients community will provide public health responses to special healthcare needs of people with rare diseases.

*Iskrov and coll.* face the financial sustainability issue, highlighting the role of health promotion, disease prevention, primary and integrated care, patient-centred care for achieving effective, appropriate and sustainable healthcare. Moreover, they remember that a proactive health workforce planning and forecasting make health systems resilient to future shocks.

*Castro and De Santis* are concerned with the integration mechanisms that are an essential condition for ensuring continuity of care and sustainability. They report on the system levers for the effective design and



implementation of integrated care frameworks, namely: political support and commitment, governance, stakeholder engagement, organisational change, leadership, collaboration and trust, workforce education and training, patient empowerment, financing and incentives, ICT infrastructure and solutions, monitoring and evaluation system.

According to *De Santis and coll.*, patient empowerment is recognised as key enabler in creating sustainability as addressing challenges faced by modern healthcare systems in terms of effectiveness, access and resilience. In the field of rare diseases, empowerment strategies have greater value due to low prevalence, lack of expertise, poor quality of life. The Authors report avenues to patient empowerment, namely: health literacy and capacity-building; shared decision-making; support to self-management.

*Jessop* considers the question of resilience and long-term sustainability that has been raised by the development of the European Reference Networks. The author refers to five “characteristics” for a resilience index adaptable to networks: awareness, diverseness, self regulation, integration, adaptation. Nevertheless, rare diseases networks should also examine their macrocul-

ture of artefacts, espoused values and unarticulated assumptions through which network members coordinate network activities.

*Montserrat and Taruscio* review the main areas for policy actions: development and implementation of European Reference Networks, as a main strategy for sharing of knowledge, clinical expertise and foster research; integration of high-quality patient registries, biobanks, and bioinformatics support, as key infrastructure tools addressing research and healthcare needs; the implementation of National Plans or National Strategies on RD in EU Member States by sharing experiences, capacity building and linking national efforts through a common strategy at a European level; actions driven by the recommendations for primary prevention of congenital anomalies (the main RD group with multifactorial aetiology); policy provisions to foster research and development of orphan drugs.

In conclusion, all of the authors enriched the debate around the sustainability of health systems for rare diseases and pinpoint the factors that are particularly relevant to match equity and an appropriate and sustainable use of resources to respond to the healthcare needs of people living with rare diseases.