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DATA AND POLICIES FOR RARE DISEASES

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Policy Brief

"HEALTH SYSTEMS FOR RARE DISEASES: FINANCIAL SUSTAINABILITY"

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INTRODUCTION

Health systems deliver preventive, diagnostic, curative and palliative health care services to people in order to improve their health. Health systems in EU Member States are varied, reflecting different societal choices. Over the last decade, European health systems have faced growing common challenges: increasing cost of health care, population ageing associated with a rise of chronic diseases and multi-morbidity leading to growing demand for health care, shortages and uneven distribution of health professionals, health inequalities and inequities in access to health care. Moreover, in recent years, the economic crisis has limited the financial resources available and thus aggravated Member States' difficulties in ensuring their health systems' sustainability. In turn, this jeopardises Member States' ability to provide universal access to good quality health care. Health systems need to be resilient: they must be able to adapt effectively to changing environments, tackling significant challenges with limited resources (1).

This is particularly true for rare diseases (2-3). These conditions are a threat to the health of EU citizens insofar as they are life-threatening or chronically debilitating disorders with a low prevalence and a high level of complexity. Rare diseases are defined as conditions affecting no more than 5 per 10 000 persons in the EU. Despite their rarity, there are so many different types of rare diseases that millions of people are affected. It is estimated that between 5 000 and 8 000 distinct rare diseases exist today, affecting between 6 % and 8 % of the population in the course of their lives. In other words, the total number of people affected by rare diseases in the EU is between 27 and 36 million. Most of them suffer from less frequently occurring diseases affecting one in 100 000 people or less. These patients are particularly isolated and vulnerable (4).

Distribution of public spending on health depends on a variety of factors, from disease burden and system priorities to organisational aspects and costs (5-6). Nowadays, virtually all health care systems face serious sustainability challenges. In the case of rare disease, priority setting involves complex value-laden choices that are often ethically controversial. This controversy arises, in part, because it involves conflicting moral obligations – beneficence versus distributive justice – which results in different levels of funding and opposing interests of a number of involved stakeholders (7). Nevertheless, EU health policy is built on principles and overarching values of universality, access to good quality care, equity and solidarity. These are of paramount importance for patients with rare diseases. Because of their low prevalence, their specificity and the high total number of people affected, rare diseases call for a global approach based on special and combined efforts to prevent significant morbidity or avoidable premature mortality, and to improve the quality of life and socioeconomic potential of affected persons (2-4, 8).

APPROACH AND RESULTS

The theoretical framework underlying the approach of this work is based upon the State of Health in the EU, a two-year initiative undertaken by the European Commission that provides policy makers, interest groups and health practitioners with factual, comparative data and insights into health and health systems in EU countries (9-10). The cycle is developed in cooperation with the Organisation for Economic Co-operation and Development and the European Observatory on Health Systems and Policies. It is also aligned to the policy objectives set out in the 2014 Commission Communication on effective, accessible and resilient health systems (1).

The 2017 State of Health in the EU report provides an in-depth analysis of EU Member States' health systems. It looks at the health of the population and important risk factors, as well as at the effectiveness, accessibility and resilience of health systems in each EU member state. Five cross-cutting conclusions come forward (10):

1. **Health promotion and disease prevention pave the way for a more effective and efficient health system**

Prevention is the key to avoid ill health and achieve a high level of mental and physical well-being effectively and efficiently. However, only a small fraction of health care budgets, political attention and stakeholder engagement are dedicated to prevention. Guided by the UN Sustainable Development Goal for 2030 to reduce by one third premature mortality from non-communicable diseases, the Commission is working closely with Member States to focus on prevention and social determinants of health (10).

2. **A strong primary care guides patients through the health system and helps avoid wasteful spending**

Strong primary care can contribute to strengthening the overall health system's performance by providing affordable and accessible care, coordinating care for patients so that they are given the most appropriate services in the right setting, and reducing avoidable hospital admissions. Efficient primary care is the key to integration and continuity between and across levels of care, which is essential for patients, particularly those with complex needs (10).

3. **Integrated care tackles a labyrinth of scattered health services to the benefit of the patient**

The rising burden of chronic disease and multimorbidity requires countries to confront the fragmentation of health services and shift towards integration: linking or coordinating providers along the continuum of care and putting the patient at the centre. Integrated care models are expected to contribute for better effectiveness, accessibility and resilience of health systems. This new challenge demonstrates also the importance of the right skill mix and training and of being able to share information effectively (10).

4. **Proactive health workforce planning and forecasting make health systems resilient to future shocks**

To strengthen prevention, primary care and integrated service delivery, health systems need a workforce of sufficient capacity and with the right skills and flexibility to meet the changing demands of health care. Yet many countries are confronted with critical health workforce problems such as supply, distribution and a traditionally oriented skill mix. Reforms in initial

education and training programmes and investment in continuous professional development are needed to foster new and appropriate skill sets (10).

5. **The patient is at the centre of the next generation of better health data for policy and practice**

More holistic, person-centred health data would have an enormous potential for improving the quality of care and the performance of health systems across the EU. Data capturing patient experiences and outcomes could markedly enrich knowledge on all topics. Generation of better information will provide a set of tools to more effectively treat patients with increasingly complex conditions and multiple morbidities, and deliver the outcomes that patients value the most (10).

IMPLICATIONS AND RECOMMENDATIONS

The 2017 State of Health in the EU report concludes that only by rethinking health systems it can be ensured that they remain fit-for-purpose and provide patient-centred care (10). Sustainability and resilience of health systems for rare diseases could be achieved by tackling the challenges identified and making the right health policy choices (2-3). Rare diseases have been one of the priorities of the Community's programmes for research and development (4). In order to improve the coordination and coherence of national, regional and local initiatives addressing rare diseases, the EU has stimulated a series of actions in the field of rare diseases. These project activities could set up in fact the practical cooperation and come up with the knowledge to translate and work on the identified five key challenges of EU Member States health systems' sustainability and resilience:

1. **Switching the focus to prevention and the social determinants of health**

Health promotion and disease prevention pave the way for a more effective and efficient health system. Aside from the unbalanced investments in prevention, social inequalities need to be tackled (10). In this context, the EUROCAT Project provided essential epidemiologic information on congenital anomalies in Europe. The project evaluated the effectiveness of primary prevention and assessed the impact of developments in prenatal screening. The outcomes of the project activity could act as an information and resource centre for the population, health professionals and managers regarding clusters or exposures or risk factors of concern, as well as could provide a ready collaborative network and infrastructure for research related to the causes and prevention of congenital anomalies and the treatment and care of affected children (11-14). The BURQOL-RD Project has addressed the social determinants of health in rare diseases in Europe. The project generated a methodological framework to measure the socio-economic burden of and health-related quality of life in rare diseases. It performed a pilot study and its outcomes could help refining and packing tools for continuous and extensive focus on the social impact of rare diseases (15-17).

2. **Guiding patients through the health system with strong primary care**

Strong primary care efficiently guides patients through the health system and helps avoid wasteful spending (10). In the context of rare diseases, newborn screening is a cost-effective approach that results in improvements in expectancy and quality of life for affected infants. This is of paramount

importance in the primary care for rare diseases, as it could eliminate the need for an otherwise expensive diagnostic odyssey, by substantially decreasing extra referrals and psychological stress for patients and caregivers (18). In 2009, the Commission launched a tender on evaluation of population newborn screening practices for rare disorders in Member States. This project activity reported on the practices of neonatal screening for rare disorders implemented in all the Member States including number of centres, estimate the number of infants screened and the number of disorders included in the newborn screening as well as reasons for the selection of these disorders. It successfully identified types of medical management and follow-up implemented in the Member States and established a network of experts analysing the information and formulating a final opinion containing recommendations on best practices and recommending a core panel of newborn screening conditions. These outcomes could help Member States in medical management and follow-up of rare disease patients, fostering primary care and avoiding unnecessary referrals (19-21).

3. Integrating care for a sustainable and effective service

Integrated care ensures that a patient receives joined-up care. It avoids the situation, where care is fragmented and patients have to search their way through a maze of care facilities. Health systems in the European Union aim to provide high-quality, cost-effective care (10). This is particularly difficult however, in cases of low-prevalence and complex rare disease. In this context, the EU established 24 European reference networks in 2017 that involve more than 900 highly-specialised healthcare units from over 300 hospitals in 26 Member States. These networks are working on a range of thematic issues, including bone disorders, childhood cancer and immunodeficiency. The European Reference Networks are virtual networks aiming to facilitate discussion on complex or rare diseases and conditions that require highly specialised treatment, and concentrated knowledge and resources. In the context of sustainability and resilience of health systems, European reference networks could help applying EU criteria to tackle rare diseases requiring specialised care, as well as ensuring the availability of treatment facilities where necessary (22-24).

4. Creating a health workforce resilient to future challenges

Proactive health workforce planning and forecasting make health systems resilient to future evolutions. Health authorities need to prepare their workforce for upcoming changes: an ageing population and multimorbidity, the need for sound recruitment policies, new skills and technical innovation (10). In the context of rare diseases, promotion and communication on the management of rare diseases by disseminating trustworthy guidelines globally is a key issue. The RARE-Bestpractices Project created a platform to improve the management of rare disease patients. The project built a comprehensive public database of trustworthy guidelines, ranging from diagnostic tests and treatments to the organization of care, to help professionals, patients, policy makers with the best and most up to date information. It also produced mechanisms to identify and prioritize rare diseases clinical research needs to optimize as well as redefine the clinical research agenda taking into consideration both patients' and clinicians' needs and interests. These outcomes are very important and could help setting up training activities targeted at key stakeholders to spread expertise and knowledge in the field of guidelines (25-27). Orphanet,

the portal for rare diseases and orphan drugs, could also greatly contribute for achieving these goals (28-30).

5. Addressing an important knowledge gap with better, patient-centred data

Patients should be at the centre of the next generation of better health data for policy and practice. The digital transformation of health and care helps capture real-world outcomes and experiences that matter to patients, with great potential for strengthening the effectiveness of health systems (10). In this context, support for registries and databases for rare diseases is crucial. The EPIRARE Project defined the state of the art of existing registries with reference to their legal basis, organizational and IT measures used, type of data collected, compliance with the rules on personal data protection, quality assurance, operational and financial support. The project agreed on a minimum data set for all rare diseases and defined criteria for quality assessment of data, data sources and procedures in the registries. These outcomes could help determining the scope, operation and governance model of rare disease registry platforms, balancing the interests of relevant stakeholders and ensuring long-term sustainability (31-33).

CONCLUSIONS

We used the five identified axes of public health challenges in the 2017 State of Health in the EU report to start a broad debate on the issue of sustainability of health care systems for rare diseases. Our analysis is suggesting a number of possible ways how the outcomes of successful EU-funded projects could help addressing and achieving sustainability and resilience of health systems for rare diseases.

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