

Sustainable public health systems for rare diseases

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Abstract

Introduction. In the framework of the Joint Action for Rare Diseases (RD-ACTION), a specific task was defined to identify mechanisms influencing sustainability, equity and resilience of health systems for rare diseases (RDs).

Method. Literature narrative review on health systems sustainability and resilience for RDs. Years: 2000-2015. Databases: PubMed, Scopus, EBSCOHost, EMBAL, PASCAL, EMBASE, STN International and GoogleScholar. Analysis: interpretive synthesis concept and thematic analysis (Dixon-Wood, *et al.*).

Results. 97 papers and 4 grey literature publications were identified. Two main topics stand out: economic evaluation and networks. The first topic did not identify widely accepted criterion to assign more weight to individuals with greater health needs. Healthcare network are identified as increasingly important for sustainability and resilience, in all of their aspects: professional "expertise", "experience" networks of users and carers; policy, learning, and interest networks.

Conclusion. Possible mechanisms for ensuring sustainability can be identified in networking, patients' empowerment and reorienting healthcare towards integrated community and home care.

Key words

- health system sustainability
- health equity
- rare diseases
- networks

INTRODUCTION

In recent years, European countries have been involved in international projects and activities on Rare Diseases (RDs) aiming to accelerate scientific research and attain the overarching goal of improving the health outcomes of people affected by these diseases. The implementation of an European Member States (EU-MSS) integrated approach to RDs calls for more comprehensive public health policies addressing the issue of sustainability. This can be achieved through a series of policy communication activities, building on other EU public health policy activities. Actually, the ongoing implementation of a better comprehensive approach to RDs is leading to the development of public health policies more appropriate. Capitalising on experience and work carried out over recent years, and with a view to further developing approaches at EU level, the Communication from the European Commission on effective, accessible and resilient health systems focuses on actions to strengthen the effectiveness of health systems, increase the accessibility of healthcare and improve the resilience of health systems. In relation to the resilience, and building on experience of recent reforms, the Commission has identified the resilience factors that helped some health systems safeguard accessible and effective healthcare services for their population, namely: stable

funding mechanisms, sound risk adjustment methods, good governance, information flows, adequate costing of health services, a health workforce of adequate capacity and with the right skills [1]. They are explained as:

- *stable funding mechanisms*: it prevents suffering from external shocks;
- *sound risk adjustment methods*: consistent system of risk adjustment and risk pooling ensure that resources are spent according to needs;
- *good governance*: strong leadership, sound accountability mechanisms and a clear organisational structure enable systems to adapt quickly to new objectives and priorities;
- *information flows in the system*: monitoring information, both at the level of individual patients or healthcare providers, enable health systems managers to make tailored, evidence-based decisions in specific sub-sectors;
- *adequate costing of health services*: the ability to cost healthcare services accurately is a prerequisite for effective decisions on investment;
- *a health workforce of adequate capacity and with the right skills*: it is essential for finding innovative solutions through organisational and technological change.

Governments around the world experience tension while setting priorities and facing innovations and sus-

tainability. This tension becomes particularly critical when resource allocation for rare diseases is concerned.

In the framework of the Joint Action for Rare Diseases (Joint RD-Action) “Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases”, a specific task on health systems sustainability was defined in order to identify and understand mechanisms that influence the sustainability, equity and resilience of health systems for rare diseases. The initial activity of the task included a literature review of the available publications on these issues. It was meant to involve policy makers, civil servants in charge of national strategies on RDs and all stakeholders in a collective responsible approach.

METHOD

To obtain an overview of sustainability and resilience of health systems for rare diseases, we performed a narrative review of the literature to select articles, documents, and reports on health systems sustainability and resilience for rare diseases.

The following health descriptors were selected: health, healthcare organization, sustainability, equity, resilience, governance, prevention, citizen/patient/families empowerment, rare diseases. The following databases were used: PubMed, Scopus, EBSCOHost, EMBAL, PASCAL, EMBASE, STN International and GoogleScholar (gray literature). The language of publication was limited to English, French, Spanish and Italian, and the year started from 2000. As a result of our search, 97 papers and 4 grey literature publications were identified.

Publications that met the inclusion criteria were content-analysed to identify the characteristics of sustainability and resilience. The analysis was primarily guided by an interpretive synthesis concept, as outlined by Dixon-Woods [2]. This mind-frame describes a process of aggregating existing concepts and approaches in the literature to come to a universal outlook. We applied a thematic analysis in this study as the guiding methodology for the synthesis of documents. This style of analysis is characterized by a clear identification of prominent themes and the subsequent re-organization of existing literature under these themes [2]. Accordingly, the characteristics of sustainability and resilience were summarized and compared with each other to identify recurring dimensions.

To further integrate the findings, the characteristics bearing the same content were clustered under a set of thematic areas and were then used to construct a framework for sustainability and resilience for rare diseases. The results of the review had been presented and discussed in a workshop with representatives of EU Member States in charge for Rare Diseases at national level, patients' organizations and professionals from public health institutions, in Rome on June 14, 2016.

The results of the workshop had been sent to all Ministries of Health representatives in charge for RDs, via email to receive additional commentary and to have ultimately validation.

RESULTS

The bibliographic search identified 97 papers and 4 grey literature publications. They were analysed accord-

ing to three main categories: sustainability, equity and resilience.

Two main topics stand out from the review: economic evaluation and networks. The first topic focuses on the available evidence on the adequacy of economic evaluation for decision-making, based upon allocative efficiency for incorporating or excluding technologies for rare diseases. There is no widely accepted criterion in the literature to weigh the expected utilities, in the sense of assigning more weight to individuals with greater health needs. Other methodologies are needed to complement cost-effectiveness studies, weighting utilities asymmetrically (that is, everyone is treated equally) [3].

As far as networks are concerned, the knowledge of people's relationships and the analysis of Social Networks in the field of public health was identified as increasingly important. Varieties of healthcare network include professional (“expertise”) networks: clinical referral “care” networks; project networks; “experience” networks of users and carers; policy networks (including policy “communities”); learning networks; and interest networks which promote particular policy or interest-group. Many health systems now use networks as governance structures. Several countries, such as United Kingdom, Netherland, Italy, United States and Australia include Patient's Organizations in health systems studies, including their sustainability. According to Sheaff, *et al.*, networks generate a “macroculture” that, on turn, generate practices and activities that can affect the community [4]. Network “macroculture” is the complex of artefacts, espoused values and unarticulated assumptions through which network members coordinate network activities. Knowledge of how network macroculture develops is therefore of value for understanding how health networks operate, how health system reforms affect them, and how networks function (and can be used) as governance structures. The study carried out by Sheaff and coll. found that in these networks, artefacts adapted to health system reform faster than espoused values did, and the latter adapted faster than basic underlying assumptions. These findings contribute to knowledge by providing empirical support for theories which hold that changes in networks' core practical activity are what stimulate changes in other aspects of network macroculture. The most powerful way of using network macroculture to manage the formation and operation of health networks therefore appears to be by focusing managerial activity on the ways in which networks produce their core artefacts.

As far as *sustainability* of health systems for RDs is concerned, the issue shares additional challenges to those faced by healthcare systems. While social and demographic pressures increase demand for health care, and technological and scientific advances continue to increase costs, in Europe, the financial crisis posed major threats to healthcare sustainability. Governments' response to the crisis was cutting public health-care budgets, and transferring healthcare costs to individuals and families through out-of-pocket payments. As a result, not only equity was jeopardized, suffering the least wealthy from the greatest health effects, but also

austerity policies affected economic growth much more adversely than previously believed. At the moment, the same international institutions imposing austerity policies are now advocating public investments to overcome economic stagnation [5, 6].

The absence of up-to-date morbidity and mortality data have clearly made the immediate effects of the crisis on health impossible to analyse, leaving policy attention focused on economic aspects. Moreover, remarkably little research has been done about the health consequences of the crisis and much of that done has been undertaken by individual researchers without additional funding [7].

The voices of key stakeholders, the health policy community, health-care managers and leaders, national governments, and politicians at both a national and a European level have not been as influential as they should be. Few studies are carried out on health policy research, on design of funding systems and their effect on financial performance, on health-care quality, and health status.

Groups concerned with biomedicine, pharmaceuticals, and medical technologies are driving the process for setting health research priorities, and government ministries of science have coordinated them nationally with little input from Ministries of Health. Research budget at EU level is dominated by biomedical topics: only 4% of the € 642 million EU cooperation programme for health research in 2011 was allocated to health systems, public health, or health policy research [8]. A report recommended that 25% of health research budgets should be spent on public health, health systems, and health policy research [8]. Horizon 2020, the biggest EU Research and Innovation programme, dedicates 38.53% of its total budget to Societal Challenges. Yet this issue concerns several fields: health, demographic change and wellbeing; food security, sustainable agriculture and forestry, marine and maritime and inland water research; Bioeconomy; Secure, clean and efficient energy; Smart, green and integrated transport; Climate action, environment, resource efficiency and raw materials; inclusive, innovative and reflective societies; security of Europe and its citizen. Horizon 2020 narrows the interpretation of health and wealth agenda, regarding health research as a lever for economic growth through exploitable intellectual property [9].

As far as *resilience* is concerned, the financial crisis created a set of economic shocks that resulted in widely varying policy responses and differing health outcomes, and thus has created the basis for future research about which policy responses can mitigate risks. Multilevel notions of resilience-*i.e.*, how individuals, communities, and entire societies positively adapt to shocks-an provide an explanatory framework that implicates the physical, psychosocial, and economic factors that help populations to resist and adapt to public health threats, such as the economic crisis [7].

Affordability and financial sustainability are the biggest issues confronting healthcare providers. Across Europe, notwithstanding the complexity and differences in how healthcare is funded and organised, the countries

face the same challenges: how to continue to provide high quality and universally accessible health services in a financially sustainable way. Healthcare expenditure is too often seen in a narrow context, purely as an economic cost. However, even at economic level, health expenditure properly organised and delivered will reduce other welfare costs, and generally improve productivity. Evidence demonstrates that significant savings can be created by investment in prevention [10, 11] and early intervention. Investment in prevention and early intervention is essential for healthcare sustainability and socioeconomic development and stability.

Fostering *empowerment* and responsibility in citizens involves assisting individuals to discover and develop the inherent capacity to be more responsible for one's own health. Healthcare systems will be more sustainable if individuals understand their rights, responsibilities, capabilities and opportunities to remain healthy and to manage their own health in the most appropriate setting, providing the political and economic context empowers them to do so. It must be acknowledged that in Europe today many people place a higher priority on their immediate survival needs. In addition, challenges arise from the fact that healthcare systems are complex, so accessing the right care at the right time in the most effective way while making the best use of health services can be a real challenge.

Strategies to empower patients take place through direct pathways, including individual effective decision making, management of disease complications, and improved health behaviours, and indirectly through strengthened support groups, empowerment of caregivers, and improved access to and efficient use of health services. Empowerment strategies require resources and capability building that should be driven by the public sector (including health and education sectors), family doctors, civil society, media, and academia. Information sharing, coupled with sustained investment in education, will also enable citizens to better understand their health condition and participate in the decision-making process to plan and manage their own healthcare plans, which will result in better outcomes.

Considering the complexity of policy-making, the WHO identified whole-of-government and whole-of-society approaches as a set of tools to manage the complex policy process and identified the following features for comparing good governance practices [12]:

- forward looking: long-term view based on statistical trends and informed predictions of the probable impact of the policy;
- innovative: questioning established methods and encouraging new ideas;
- informed by evidence: using the best available evidence from a range of sources;
- inclusive: taking account of the impact of the policy on the needs of everyone directly or indirectly affected;
- joined-up: horizontal and vertical integration;
- adaptive: learning from experience of what works and what does not;
- evaluative: including systematic evaluation;
- accountable: being democratically legitimized, trans-

parent and responsive to the demands of citizens.

Many existing whole-of-government and whole-of-society approaches focus on *communication, cooperation and coordination*. The final step in whole-of-government approaches, collaboration or even integration, in which risks, responsibilities and rewards for a common goal are shared, seems to be less common and the most difficult to achieve [1].

While many whole-of-government applications have led to a strengthening of central coordination bodies, it may be that the cooperation required in whole-of-government approaches works best at lower levels of governance, such as local authorities.

Whole-of-government approaches at this level can significantly enhance transparency, accessibility and responsiveness, as long as institutional arrangements are adaptive to change and create accountability to the citizens they serve. At local level, however, the move from a whole-of-government to a whole-of-society approach through the involvement of many local stakeholders has become a strong feature of “smart governance”.

Despite the centrality of *equity* in policy, research review highlights that patients’ access or use of healthcare services is inequitable. However, while there is an implicit assumption of inequity, there is little evidence to support this assumption, as studies rarely assess need or demand for services rather than patterns of utilization. Moreover, the studies are biased towards issues of accountability and deserve little attention to legitimacy issues, that concern normative aspects of fairness and equity. The real political, economic and ethical question is the source of the required financing for health services. Very rich countries can still afford to rely largely on private health insurance or out-of-pocket payment, but this option is seriously hampering equity. Moreover, an economic system that is concentrating wealth in the hands of a few, calls for political considerations about redistribution of resources and wealth.

DISCUSSION AND POSSIBLE INITIATIVES

Aluttis *et al.* [14] identified public health capacity dimensions in the organizational structure, resources, partnership, workforce, knowledge development, leadership and governance and country specific context with relevance for public health.

Health systems sustainability shares several dimensions with WHO/EURO’s Essential Public Health Operations [14] and with the public health capacity dimensions identified by Aluttis *et al.* [15], namely organizational structure, resources, partnership, workforce, knowledge development, leadership and governance and country specific context with relevance for public health. More specifically, the organizational structure shapes the infrastructural ability of the system to contribute to the goals to healthcare systems for RDs. Allocation and provision of human and financial resources to healthcare systems for RDs; collaboration among organizations for effective practice; qualified human resources with adequate skills and knowledge; knowledge development as a base providing information on the health status and supporting evidence-based health policy and interventions at all levels; leadership and

governance as ability and willingness of governments to improve public health by developing and implementing effective health policies and by expressing qualities in leadership and strategic thinking; all of them represent key elements and mechanisms for the sustainability of the health systems for rare diseases. Moreover, the political context and other characteristics of a country may have an influence on health policies and capacity building efforts with relevance for RDs.

A European Steering Group on Sustainable Healthcare identified initiatives for sustainable healthcare [16]. According to our literature analysis, a few of the initiatives identified by the European Steering Group on Sustainable Healthcare could be adapted to healthcare for rare diseases. They address Member States and Ministries and concern 1) prevention; 2) empowerment and resilience; 3) integrated care based on patients’ pathways and care delivery shifted to community and homes. In particular these initiatives suggest to focus respectively on:

Prevention

- Governments: to consider health and equity aspects in all policies;
- Government ministries: to develop joint budgeting mechanisms between ministries and intersectoral action, thus addressing the current silo approach¹ (e.g. health and social affairs, education, and economic and budget ministries).
- Member States: to strengthen registries, and develop practice guidelines on systematic data collection and surveillance, so as to better inform national prevention and treatment strategies.

Empowerment and resilience

- Member States: to fully transpose the Directive on the application of patients’ rights in cross-border healthcare to improve citizens’ access to information on healthcare systems and develop and implement information and education programmes for citizens on care available;
- National Ministers of Education: in the context of prevention campaigns, to integrate early education programmes in school curricula addressing health determinants at early stages in life;
- Ministries of Health and Education: to jointly develop programmes aimed at increasing the level of health literacy among the general population;
- Member States: to foster the development of multidisciplinary partnership and comprehensive approaches between policymakers, healthcare providers, community and health planners, patients and pharmaceutical companies to address issues with regards to rare diseases.

¹ A silo mentality can occur when a team or department shares common tasks but derives their power and status from their group. They are less likely to share resources or ideas with other groups. Collaboration in a business culture with silos among teams or departments will be limited, unless collaboration benefits the members of the department. In addition, the members of a silo tend to think alike. They get their power from association with their function.

Integrated care based on patients' pathways and care delivery shifted to community and homes

- Member States and European Commission: to create a platform of exchange for European and non-European countries' experiences in establishing healthcare systems fit to address chronic care and rare diseases;
- Member States: to develop guidelines and funding mechanisms to develop community and home care;
- Member States: to develop integrated care models for rare diseases which efficiently link and leverage rare disease registries;
- Member States' governments and healthcare providers: to conduct frequent performance audits in health services to identify opportunities for efficiencies, thereby improving their performance and outcome.

Finally, better organisation and management of healthcare for RDs is expected not only to decrease economic burden but also to enhance quality of life, ensuring both sustainability and equity to healthcare systems for RDs. Centres of expertise and reference networks are a key public health policy tool to address health inequalities due to rare diseases in the EU.

CONCLUSIONS

Sustainability of health systems for RDs shares additional challenges to those faced by health systems, that are stressed by the current financial crisis. Many similarities exist between public health capacity and sustainability for RDs, and possible mechanisms for ensuring sustainability can be identified, namely:

- networking;
- patients' empowerment;
- reorientation of healthcare towards integrated community and home care.

Both networking and patients' empowerment rely upon community and health systems' resilience, *i.e.* how individuals, communities, and entire societies positively adapt to challenges. The active role of patients' representatives is now recognised as a major contribution to catalyzing cooperation and sustainable

development, as well as in its political role of raising awareness and advocating at national, EU and international level.

According to WHO, the following principles should be taken into account for complex policy processes:

- identify several initial domains for action as lever points for change;
- assemble around each lever point a strategic network of key stakeholders from government and civil society;
- bring together all the networks created according to the lever points within a whole-of-society compact that is supported by a platform to share information and research and build capacity.

WHO identified two sets of tools to manage the complex policy process: a whole-of-government and a whole-of-society approach. Both of them are fundamental for networking and must be adapted to each country's unique circumstances and background.

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Conflict of interest statement

None of the authors has any kind of conflict of interest to declare.

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