



EURORDIS

European Organisation for Rare Diseases

EURORDIS Position Paper

on Commissioner Byrne's reflection document for a new EU Health Strategy

" Enabling Good Health for all"

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The Position Paper has been endorsed by EURORDIS Board of Directors.

Eurordis, the European Organisation of Rare Diseases, is a patient-driven pan-European network of more than 200 rare diseases associations from 21 countries, 17 of which are EU member states, including ten National Rare Diseases Alliances. Thereby, Eurordis reflects the voice of an estimated 25 to 30 million citizens currently affected by 5000 to 8000 rare diseases in the enlarged EU.

What are the characteristics of rare diseases?

First of all their rarity, their low prevalence (less than 1/2000). They are severe, life-threatening and chronically debilitating. They are heterogeneous. They evolve, with acute and non-acute phases. To cure them, there is no treatment, except in very few cases. They are often difficult to diagnose. There is a lack of knowledge, lack of information, lack of training and experience of physicians, lack of centres of care for rare disease. Therefore, patients and patients groups are isolated. The day-to-day specific technical care changes over time, and is long term. Patients take care of themselves, when they can, or relatives act as carers. There is a lack of social recognition and public awareness. Patients living with rare diseases do not constitute a critical mass for the health care system. There is no specific public health policy for patients living with rare diseases. Nor is there a research policy or a market for drug development, devices and services. This is the "orphan patient" situation.

We very much welcome David Byrne's initiative to launch a reflection process on what the EU needs to be doing in order to achieve good health for all across Europe. We are pleased to take this opportunity to provide input as part of the process leading to a future EU health strategy.

Our contribution will be made up of two parts: firstly, the aspects that raise some concerns for people living with rare diseases and towards which we have a critical position, and, secondly, the elements that we strongly support and that we wish to emphasise even more.

1. The aspects raising some concerns for 30 million people living with rare diseases:

In order to achieve a healthy Europe, all citizens across the EU have to be equally protected and provided with an even level of health care, whether they are currently in good health or affected by a common or a rare disease.

We believe that this document addresses only a part of existing causes of ill health: the preventable, known, treatable diseases. On the contrary, rare, genetic, chronic, non-preventable diseases do not seem to find an appropriate place in the proposed framework of reflection.

Primary prevention is indeed an important tool for promoting good health, but the major limitation of this tool - if considered as the only means to achieve good health - is that it can be applied to avoid only certain diseases, namely the ones whose causes, development and evolution are known and preventable.

This document therefore raises some concerns as it focuses mainly on primary prevention and would lead to the establishment of a health system that would prove inappropriate to meet the needs of a large part of the EU population. Far from reducing inequalities across the EU, the overall proposed perspective would widen the disparities between citizens according to which kind of disease affects them.

Sentences such as "Europe should take action to avoid ill health in the first place" (p.2), "European citizens need reliable and user friendly information about how to stay healthy and the effects of lifestyle on health" (p.3) are appropriate when tackling preventable diseases linked to alcohol, nutrition, tobacco, behavioural, social and environmental factors (such as p.7 et 8). But Europe cannot take action only in this area, it would simply leave the 25 to 30 million people affected by rare diseases outside the scope of EU Health Policy and would not "achieve good health for all across the EU".

Most rare diseases (80%) have a genetic origin and cannot be prevented by right choices of life-style. The remaining 20% have other origins such as viruses, bacteria, or cell proliferation. Patients living

with rare diseases would be excluded from the proposed health perspective if the document is not re-balanced between primary prevention, on the one hand, and secondary (early diagnosis by screening) and tertiary prevention (care and treatment to prevent health debilitation and clinical manifestations of the disease), on the other hand.

The different levels of prevention--primary, secondary and tertiary--are important for rare diseases in many ways :

- Genetic counselling for families having a history of rare disease. This tool should be developed and properly organised: genetic testing, pre-implantation diagnosis, pre-natal diagnosis ;
- Early diagnosis for a better care and management of the disease in order to avoid mistakes and long suffering ;
- Management of the disease in order to avoid - or slow down - the consequences of an incurable disease (physiotherapy, surgery, ventilation, nutrition, medical devices, technical aids, etc). Knowing that there is often no cure for rare diseases, this kind of prevention and its access for all patients (including exchange of good practices) should be encouraged within the EU Health Strategy.

We also believe that the document should mention more explicitly the need to improve, within the 7th EU Framework Programme for research, the funding dedicated to rare diseases with the aim to foster innovation, increase knowledge and develop the best treatments.

2. The aspects we do support and wish to emphasise:

- At the individual level: we know from direct experience how much ill health is a heavy financial burden as rare diseases have a tremendous financial impact on the family of the patients. Treatments for rare diseases are very expensive, and generally not covered by the national public health system. Paying carers to help in assisting patients is not affordable for every family, so usually one parent has to stop working completely to take care of a sick child. So, on the one hand, expenses rise dramatically and on the other, the income decreases. This leads to real pauperisation of the families and losses in purchasing power, which increases even further their exclusion.
- At EU level: improving treatments and health assistance would save a lot of money, allowing the rare diseases patients who can work to attend work more regularly, avoiding depression and boosting productivity. We therefore strongly support the idea that "each health euro better spent could make a net saving both for individual well-being and for EU economic competitiveness" and that "good health is key to economic growth" (p.2). "Improving health must become an economic priority" and "health expenditures have to be seen as long-term investments and as a key driver of economic growth" (p.4). And also: "Europe needs a paradigm shift from seeing health expenditure as a cost to seeing effective health policies as an investment" (p.6).
- Good Health as a shared responsibility: "The EU must achieve synergies with national authorities, stakeholders, international organisations and foster cooperation between Member States". This statement is precisely best illustrated in the field of rare diseases, where parallel initiatives are scarce in other European or international fora (contrary to many other health agendas, where e.g. the WHO has established relevant structures, and where duplication of effort would easily occur). Synergies at EU level would help achieve Patient Mobility, Second Opinion, Diagnosis and Treatment Centres of Reference and Networks. Rare diseases have to be tackled at the European level, because of the small number of people affected by each disease at national levels and the limited number of researchers working on these pathologies. "There are some areas where synergies and savings can be achieved, such as exploiting European centres of expertise and exchanging knowledge on issues such as quality improvement and assessment of health technologies" (p.4). This is fundamental in the field of rare diseases where the already limited knowledge is often scattered throughout the EU: it would save money and avoid duplicating the testing of treatments. A good instrument to achieve this goal would be increasing the funding for these activities within the 7th Framework Programme for Research.

- Innovation and access to patients: “The health sector is driven by scientific and technological progress. Everybody wants and expects access to the latest and best treatment” (p.5). Innovative treatments have to be available as quickly as possible and accessible to all. This could be facilitated by the creation of new health research structures in Europe (p.9) and by a European health innovation powerhouse channelling research to new medical appliances and medicines and disseminating results across the EU”, as stated on page 10, but also by developing an active European pharmaceutical industry in areas such as Orphan Drugs where 250 new orphan drugs have been designated in 4 years.
- Quality and safety: “The Constitution gives the EU the role of setting quality and safety standards for medical products and devices” (p.3). Often it is thought that this quality and safety concern only relates to the new Member States but it has to be underlined that, in practice, there are differences in this field according to whether you deal - within EU15 - with a common or a rare disease. The latter have neither protocol, nor suitable updated treatments. We would like some reassurances concerning how this will be implemented for rare diseases, because currently equality does not exist in this field. Concerning health quality assessment, diagnostic tools such as gene testing and biochemical tests would lead to the creation of Centres of Reference. The accreditation of Centres of Reference at the EU level is requested by patients organisations, health professionals and national health authorities.
- Focusing more on the citizen: “Involve citizens in policy-making” (p.4). “Openness and civil society participation, two core principles of good governance”. “Supporting networking of patient's organisations” (p.9). Of course we couldn't agree more. It has to be underlined that patients organisations of rare diseases have developed a real expertise and are of fundamental importance for the patients, as they are the only ones being able to reach small groups of isolated citizens who need appropriate support and relevant information they do not find anywhere else. To make patients organisations' involvement effective, certain conditions have to be fulfilled: a funding mechanism has to be established in order to develop information-to-patients groups, training and networking activities, and patients organisations have to be involved at the very first stages of the decision-making process.

In conclusion:

We urge the Commission to include in this reflection document a paragraph where rare diseases are explicitly recognised with their specificities. Rare diseases could represent a good model for a future more integrated Pan-European Health Policy.

The vision of a future EU united for health, linking research centres, exchanging data, where easy and prompt access is secured for all, where there is a high equal level of health care and no trouble finding clear and reliable information, with the very best European Centres of Reference, is the best possible goal to be achieved.

We are keen to see how favourable conditions to reach this goal will be put in place in the EU and how rare diseases will be taken into account in the shaping and implementation of the future EU Health Policy Strategy.