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COMMISSION STAFF WORKING DOCUMENT

Summary of the Impact Assessment
Accompanying the
COMMUNICATION FROM THE COMMISSION TO THE COUNCIL, THE EUROPEAN PARLIAMENT, THE EUROPEAN ECONOMIC AND SOCIAL COMMITTEE AND THE COMMITTEE OF THE REGIONS
On
Rare Diseases: Europe's challenges

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1. INTRODUCTION

Rare diseases are life-threatening or chronically debilitating diseases with a low prevalence and a high level of complexity. Most of them are genetic diseases, the others being rare cancers, autoimmune diseases, congenital malformations, toxic and infectious diseases, among other categories. They call for a global approach based on specific and combined efforts to prevent significant morbidity or avoidable premature mortality, and to improve quality of life or socio-economic potential of affected persons.

The prevalence for a rare disease is currently defined as affecting no more than 5 per 10 000 persons in the European Union. Whilst this prevalence rate seems low, it translates into approximately 246 000 persons per disease in the EU 27 Member States (MS). Based on present scientific knowledge, there are between 5 000 and 8 000 distinct rare diseases that affect up to 6% of the total EU population at one point in life. In other words, this equates to between 29 and 36 million people in the 27 MS that are affected, or will be affected, by a rare disease.

2. PROBLEM DEFINITION

2.1. Lack of Recognition and Visibility of Rare Diseases

Although rare diseases heavily contribute to morbidity and mortality, they are mostly invisible in health care information systems due to the lack of appropriate coding and classification systems. The lack of formal identification in health systems thus imposes medical and financial barriers to receiving treatment for an unrecognised disease that consequently lacks allocated funds and resources, thus creating a cycle that maintains the current inefficiency. Furthermore, misdiagnosis and non-diagnosis are the main hurdles to improving life-quality for thousands of rare disease patients.

2.2. Lack of Policies on Rare Diseases in the Member States

Within the Member States, there is fragmentation of the limited resources available for rare diseases, thus it is essential to have a specific plan to concentrate and make efficient use of these resources that would otherwise fall below the threshold for efficacy. The lack of specific health policies for rare diseases and the scarcity of the expertise, translate into delayed diagnosis and difficult access to care.

2.3. Lack of Effective Healthcare, Research, and Regulation for Rare Diseases in Europe

2.3.1. Inequitable Access to Expert Healthcare

There is a lack of reference networks and access to care, resources, and expertise that may well only be available in another Member State.

2.3.2. Fragmented Research

There is a very close link between research and the possibilities for diagnosis and treatment of rare diseases. Therefore, further research on rare diseases is needed but is hampered by inefficiency and fragmentation of the limited resources available.

2.3.3. Insufficient Legislative Framework

The current EU legislative framework is poorly adapted to rare diseases. Relevant existing Community legislation, for example on clinical trials and marketing authorisation of medicinal products, is proving unsuitable and insufficient when applied to rare diseases.

2.4. Subsidiarity

There is probably no other area in public health in which 27 national approaches could be considered as inefficient and ineffective as with rare diseases. The reduced number of patients for these diseases and the need to mobilise resources means the scale and nature of effective action requires action at European level, in accordance with Article 152 of the Treaty establishing the European Community.

It is not feasible to have a centre for every disease in every MS due to the high levels of resources that would be required. The idea is that the expertise, rather than the patients, should travel – although patients should also be able to travel to the centres if they need to.

2.4.1. Necessity Test

Member States have the prime responsibility for protecting and improving the health of their citizens. As part of that responsibility, it is for them to decide on the organisation and delivery of health services and medical care to patients suffering from a rare disease. However, the fundamental aims of the EU in terms of free movement of patients, equitable recognition of diseases, and equitable access to safe and efficient orphan drugs or cooperative research on rare diseases, necessarily have an EU health dimension.

A key reason for taking action now on rare diseases is the current revision of the International Classification of Diseases (ICD). The new ICD-11 also aims to include rare diseases and to do this effectively from a European perspective there needs to be a central coordinating point.

2.4.2. Added-Value Test

The EU can add value through a wide range of activities. These include working to reach critical mass or obtain economies of scale—for example sharing information on rare diseases where only a small number people are affected in each Member State—or performing collaborative multidisciplinary research, which proves the most efficient way to better understand the diseases and develop preventive, diagnostic and therapeutic methods. Clear added-value examples can be identified in the following four areas:

- Reducing Inequities in Health in the EU
- Creating a Coherent Framework for Identification of Rare Diseases and Europe-wide information sharing;
- Added-value of a new EU approach on rare diseases, improving information, identification and knowledge on rare diseases to set a strong basis for diagnosis and care of patients;
- Creating an Improved Framework for Research on Rare Diseases

3. OBJECTIVES

The overall objective for Community action on rare diseases is to support Member States in ensuring effective and efficient recognition, prevention, diagnosis, treatment, care, and research for rare diseases. This is supported by the Commission's strategic goals of prosperity, solidarity, and security. This is to be achieved through three specific objectives.

3.1. Improving Recognition and Visibility on Rare Diseases

The key to improving overall strategies for rare diseases is to ensure that they are recognised, so that all the other linked actions can follow appropriately. The EU should cooperate closely with the WHO in the process of revising the existing ICD in order to ensure a better codification and classification of rare diseases.

3.2. Supporting Policies on Rare Diseases in the Member States

Efficient and effective action for rare diseases depends on a coherent overall strategy for rare diseases mobilising scarce and scattered resources in an integrated and well-recognised way, and integrated into a common European effort. That common European effort itself also depends on a common approach to work on rare diseases across the EU, in order to establish a shared basis for collaboration.

3.3. Developing European Cooperation, Coordination, and Regulation for Rare Diseases

The Community should aim to better coordinate the policies and initiatives at EU-level, and to strengthen the cooperation between EU programmes, in order to maximise further the resources available for rare diseases at Community level, in particular to ensure:

– effective coordination of research and technological development;

– access to appropriate expert healthcare to, as well as specialised and adapted social services for rare disease patients;

– and adaptation of the framework of legislation and action at Community level to the specific needs of rare disease.

4. POLICY OPTIONS

4.1. Baseline Option

Continuing with project-based work without a European reference point within current legal framework

Under this option, the Commission would continue to support individual projects aiming to improve the recognition and visibility of rare diseases, without providing formal guidance or recommendation to Member States regarding how to ensure efficient and effective strategies.

4.2. Commission Communication and Proposal for a Council Recommendation

Under this option, the Commission would provide a formal statement of the definition of rare diseases within the EU, and set out its intentions for recognition and visibility of rare diseases at European and global level and set out an overall strategy for European work on rare diseases.

The Commission would also propose a Recommendation of the Council, recommending that Member States establish coherent and comprehensive national strategies for rare diseases.

4.3. Re-establish Formal Rare Diseases Programme

Under this option, the Commission would propose establishing a specific programme with a single detailed strategy for rare diseases healthcare at Community level. The programme would be established under Article 152 of the Treaty, in order to take forward specific projects on rare diseases in a similar way to the previous specific programme on rare diseases. The Commission could also adopt measures under the Statistical Regulation of the European Parliament and of the Council on Community statistics on public health and health and safety at work in order to put in place a binding legal requirement for the collection of data on rare diseases by the Member States.

5. ANALYSIS OF IMPACTS

5.1. Social Impacts

Given the complexity and time-consuming nature of establishing national strategies from scratch in the Baseline Option, it seems unlikely that without providing a clear reference point bringing

together existing best practices from across the Union, Member States would be able to establish such strategies. This could lead to even greater inequities developing between the Member States.

A Council Recommendation would provide a formal legal and political commitment to the Member States whilst maintaining flexibility in the implementation. This approach is specifically provided for in Article 152 as an appropriate tool in the health area, balancing effective guidance and shared commitment with respect for subsidiarity. This would lead to greater equity and quality in the provision and access of services, and thus have a positive effect on the health of the population within the Member States.

The re-establishment of a formal rare disease programme would offer very little increase in the efficiency of actions compared to a Commission strategy. Thus, re-establishing a formal programme would not offer significant advantages over the other options outlined.

5.2. Environmental Impact

Due to the nature of the initiative, the environmental impact is negligible, and will not be considered further.

5.3. Economic Impacts

Successful intervention on rare diseases could also have economic impact in improving efficiency and effectiveness in the use of resources for rare diseases. The establishment of the French multi-annual (2005-2008) strategy for rare diseases will cost €86.66m with a further €20m to be spent on research. The budgetary consequences for public authorities in establishing these strategies without guidance and a European approach make the Baseline Option nonviable for many Member States.

The technical work for Council Recommendation option can be taken forward with support from the existing health programme, and by centralising efforts, which will be more efficient and less burdensome for national health systems and public authorities. Improving the efficiency of action to address rare diseases will bring significant benefits both for the individual patients and for the efficient use of resources for health systems overall. Given the non-binding nature of the initiative, the likely impacts are not expected to be burdensome to any group or sector.

The administrative burden on public authorities of requiring data for the 5 000-8 000 rare diseases from throughout the Union, as proposed in the third option, would be substantial. The additional cost of integrating data collection on rare diseases into the European statistical system would also be substantial. This option would also raise questions about subsidiarity, given the differences in organisation and delivery of health services and medical care throughout the Union. Although areas such as research and technological development would benefit, to re-establish a formal EU programme on rare diseases would require a substantial level of funding to be viable, therefore this does not appear to be the most efficient approach.

6. COMPARING THE OPTIONS

6.1. Improving Recognition and Visibility of Rare Diseases

	Baseline Option	**Commission Communication**	**Compulsory Requirement for Data Collection**
Advantages	Better identification & categorisation	Improved recognition; engagement of relevant stakeholders; adoption of the results.	Improved evidence base; improved public health monitoring; improved & more

			equitable services provision.
Disadvantages	Reduced likelihood of classifications being adopted; duplication & inefficiency of work; continued inequities in access to care.	Depends on collaboration of a wide range of stakeholders to succeed.	High administrative burden; high cost of integration into statistical system; disproportionate level of action.

6.2. Supporting Policies on Rare Diseases in the Member States

	Baseline Option	**Council Recommendation**	**EU-Level Healthcare Strategy for RD**
Advantages	Maximum flexibility for Member States to organise health systems as they wish.	Formal legal & political commitment; maintain flexibility; increased efficiency & efficacy of actions; pooling of resources.	Detailed guidance at EU-level; more effective in detailing best practice; increased healthcare provision.
Disadvantages	Inequities in access & quality of healthcare persist; lack of clear reference point; inefficient establishment of national strategies; resources remain fragmented.	No legal requirement for Member States to comply.	Significant restructuring of national health systems; issues with subsidiarity.

6.3. Developing European Cooperation, Coordination, and Regulation for Rare Diseases

	Baseline Option	**Commission Communication**	**Re-establish Rare Diseases Programme**
Advantages	Avoids any need for redirection of existing Community actions.	Improve equity in access to & quality of healthcare provision; enhance cross-border cooperation; decrease in mortality & morbidity; reduce inefficiencies; stimulate research; facilitated introduction of technology.	Provides political visibility of Community funding.
Disadvantages	Continuing actions inefficient; lead to greater inequities; resources remain limited and scattered.	Depends on cooperation across a wide range of programmes and actors	Substantial level of funding required (not available under existing financial perspectives); lack of

		at Community level.	integration in other policy areas; inefficient approach.

6.4. Summary

On this basis, the preferred option is to bring forward proposals for a Community strategy for rare diseases set out in a Commission Communication, with a shared commitment to be sought through an accompanying proposal for a Recommendation of the Council on establishment of coherent and comprehensive strategies for rare diseases based on Article 152 TEC.

7. MONITORING AND EVALUATION

7.1. Data Collection

A Data Set for Rare Diseases Indicators will be established based on the ongoing works of the technical support structures. The Data Set would cover the following areas (an indicative, non-comprehensive list only):

- Demography, Epidemiology, and Health Status
- Determinants of Health and Socio-economic Factors
- Health Services
- Research and Technology Development
- Equity, Regional Differences, and EU Initiatives

7.2. Comitology and Monitoring Mechanism

An EU Advisory Committee on Rare Disease (EUACRD) would be created in order to accomplish the tasks currently performed by the EU Rare Disease Task Force. The future EUACRD shall be composed of representatives of the 27 MS, incorporating experts from the Health Programme and FP Projects, representatives of the patient's organisations, representatives from industry, and other interested bodies.