Response to the European Commission Public Consultation on rare diseases: Europe’s challenges

The European Haemophilia Consortium\(^1\) (EHC) is a European patient group representing national member organisations from 44 countries in Europe including members in all 27 EU Member States and the 2 candidate countries Croatia and Turkey.

Haemophilia\(^2\) is an inherited lifelong bleeding disorder that prevents blood from clotting properly and is classified as a rare disease (as it affects less than 5 per 10,000 persons in the European Union). The comprehensive treatment and care of this rare disease requires an excellent medical expertise as well as a ready access to a range of services provided by a multidisciplinary team of specialists.

The EHC is working to reduce the burden of the disease on both the individual and society. Its mission is to improve the quality of life of people with Haemophilia and their families in Europe.

Introduction

Considering the challenge of developing comprehensive care and treatment for rare diseases such as haemophilia in Europe, EHC welcomes the initiative of the European Commission to launch a public consultation on a Community action in the Rare Diseases (RD) field.

We are aware of the limitations drawn by the subsidiarity principle at the EU level and by Article 152 of the EC Treaty but we believe that it is important for the forthcoming EU communication on community action to be endorsed by the Council of Ministers. This should result in a recommendation which would send a strong political signal to European society, patients and their families on the Community willingness to act in the area of Rare Diseases at the European level in a coordinated way.

We agree that patients suffering from a rare disease such as haemophilia should have the right to equal prevention, diagnosis and treatment like any other patients and believe that the European Union and its member states should give themselves the means of this policy.

\(^1\) For more information on the EHC, please go to www.ehc.eu
\(^2\) Haemophilia is primarily a genetic disorder, affecting almost exclusively males. People with haemophilia do not have enough or no clotting factor, a protein in the blood that is necessary to control bleedings. The severity of a person’s bleeding disorder depends on the remaining clotting activity of the factor that is lacking. There are three levels of haemophilia: mild, moderate and severe.
We are aware of the different initiatives taken by the European Commission in the field of rare diseases, including the set up of a task force on rare diseases but a lot remains to be done for better patient welfare and we hope that the initiatives proposed by the Commission in this communication will be followed by concrete action.

**Question 1: Is the current definition of rare disease satisfactory?**

“Less than 5 per 10,000” is the core element of the definition adopted by the Community action programme on RD (1999-2003). We believe that this is an acceptable definition.

However, the EHC would like to stress that the definition of RD should not only focus on the question of prevalence but should also insist on the need for a global approach to the treatment and care of these diseases to manage all their consequences for the patients and their families, prevent mortality and improve the quality of life and socio economic opportunities.

**Question 2: Classification and codification of RD**

Haemophilia and other coagulation disorders are classified from code 286.0 to code 286.9 according to ICD9-CM. We agree that the improvement of the coding and classification in the area of Rare Diseases should be a priority, especially at a time when the existing International Classification of Diseases (ICD) is revised by the World Health Organisation.

**Question 3: For a European Inventory of RD**

The EHC is in favour of the establishment of an inventory of rare diseases to maximise awareness. Such an initiative should be supported by the European Commission, including through financial support. This would of course help national and regional systems to better deal with rare diseases.

This theme is strongly related to the need for data collection in the field of rare diseases, including for Haemophilia.

The EHC has long been lobbying for the establishment of compatible national registries across the EU to collect useful data on patients suffering from Haemophilia.

The EHC strongly believes that the widespread development of compatible registries for a disease such as Haemophilia would help national and regional health care systems to offer more comprehensive care to patients.

European compatible national patient registries will allow for better resource planning and allocation, provide accurate data on patient numbers, geographical

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spread and adverse events such as the incidence of inhibitors. They can help identify changing care needs and improvements, and measure the progress required for better haemophilia treatment. These registries can form the basis of an effective surveillance system for monitoring the safety of treatments and collect data on a long term basis (to assess, for instance, conditions in which patients suffering from haemophilia age). They can also support benchmarking activities between Reference Centres in Europe by measuring their performance in different areas of disease management.

**Question 4: Should the European Reference Networks privilege the transfer of knowledge? The mobility of patients? Both? How?**

European Reference Networks should privilege the transfer of knowledge, using electronic tools.

As far as haemophilia is concerned, patient associations are well connected in Europe and information on medical and scientific issues is effectively disseminated.

Nevertheless, the EHC welcomes the establishment of the European Association for Haemophilia and Allied Disorders (EAHAD), a European transnational medical body, that aims to ensure the provision of the highest quality of care available and to advance the education of medical practitioners, clinical scientists, professions allied to medicine and the general public in the knowledge of haemophilia and allied disorders and their treatment.

EHC and EAHAD are partners in the European Haemophilia Safety Surveillance System (EUHASS), a project financed by DG SANCO. The objective of EUHASS is to set up and maintain a database of all haemophilia centres in Europe as well as a Health Information System for monitoring and wide communication of health information and treatment safety. The further development of EUHASS has the potential of providing the basis for a European Haemophilia Reference Network. Across the network, Centres of Excellence focussing on different issues (e.g. inhibitor management, orthopedic complications, genetic counselling, etc.) could be identified on the basis of their observed and measured performances. Centres of excellence can provide scope for a more appropriate use of resources, concentration of knowledge and expertise and high quality standards of care.

EHC also welcomes the development of the European Network of Rare Bleeding Disorders with the Universita degli Studi di Milano, Italy, as Project Leader. 16 European states (and amongst them 11 EU member states) are part of this network, but more states and centres should join this network. The aim is to develop a network of European centres dealing with patients affected by rare bleeding disorders, in order to collect information but also to test the operability of a European network using a common model for data collection, query and report.

The existence of these networks as well as of others (cystic fibrosis, dysmorphology, etc) need to be widely publicised to ensure the widest scope possible and a good information-sharing process. This is one of the reasons why EHC welcomes the contribution of Eurordis to the debate at a European scale and
is grateful for its role in facilitating information dissemination and exchange. We recognise Eurordis as a privileged partner.

The European Network of Rare Bleeding Disorders is an example of added opportunities offered by the cooperation of countries within the EU in terms of research, information exchange and professional training.

By building a European Haemophilia Reference Network and by facing the legal and economic issues that impede patient mobility across the enlarged EU, a step could be taken to cope with the wide disparities that affect haemophilia care. The organisation of a consensus conference with the support of the European Commission and Member States, on the issue of haemophilia treatment would also help find some common grounds and improve the treatment of this disease in each country of the enlarged EU.

**Question 5: Should on-line and electronic tools be implemented in this area?**

On-line and electronic tools have of course their role to play in the development of a knowledge and information sharing process for rare diseases.

However, the development of these tools and the selection of data collected or disseminated, via the Internet for instance, need to be carried out in dialogue with patient organisations, experts, and healthcare professionals to identify the most relevant issues. **Harmonising** this data collection and information sharing at the EU level should be a priority.

Opportunities offered by telemedicine in haemophilia diagnosis and treatment (as well as for the wider issue of rare diseases) should be investigated further and supported as a part of Reference Centres networking.

**Question 6: What can be done to further improve access to quality testing for RD?**

Genetic screening and counselling are very important for rare diseases such as Haemophilia and the improvement of the screening techniques will greatly increase early diagnosis rates. The exchange of information across European countries and the definition of very good EU agreed standards and procedures will be essential to better access quality testing for RD such as haemophilia, especially in new Member States.

In the specific case of haemophilia, quality testing means that diagnostic tools allow doctors to determine the degree of gravity of the disease (i.e. mild, moderate or severe) with accuracy. This is not currently the case in some countries across Europe and the development of EU agreed standards for coagulation laboratories could be a good way to remedy to this challenge and refine diagnosis. To guarantee such standards in the most efficient way, the transnational external quality assurance of laboratories should be encouraged and adequately financed.
Question 7: Do you see a major need in having an EU level assessment of potential population screening for RD?

An assessment, at the EU level, of potential population screening for rare diseases would be an excellent initiative to ensure cooperation between Member States in the area of population screening and the development of a harmonised approach in this field.

For the diagnostic of haemophilia, genetic counselling and screening, especially towards women as potential carriers, is extremely important for families already affected by this genetic disease.

EU-level assessment for haemophilia

An evaluation of screening for haemophilia across the EU to study the approach of the different member states on this issue could be, for instance, used as a good case study for Rare Diseases in general. It would allow, amongst other things, taking a careful look at the link (or absence of) between the comprehensive and effective treatment of haemophilia and the increase or decrease of requests for pre implantation genetic diagnosis.

Question 8: Do you envisage the solution to the orphan drugs accessibility problem on a national scale or on an EU scale?

Networks of care

The EHC would like to encourage networks of care suppliers at EU level to combine experiences and to better manage unexpected complications.

Orphan drugs accessibility

The EHC supports the regular reporting by the European Commission on the progress made to identify bottlenecks for the access to orphan drugs.

The problem of accessibility to orphan drugs should be tackled on a national and EU scale. Patients suffering from rare bleeding disorders would for instance greatly benefit from better accessibility to orphan drugs and both the EU and national member states have to show their willingness to solve accessibility problems.

The EHC particularly welcomes any initiative of the EC aimed at promoting policies (e.g. dual track prices, transnational consortium) to increase the supply and availability of costly haemophilia treatment in European countries with lower GDP.

Question 9: Should the EU have an orphan regulation on medical devices and diagnostics?

We do agree on the need for an orphan regulation on medical devices and diagnostics.
The EHC believes that it is important to support the efforts of the industry in the development of new medicinal products – but also the improvement of the anti-haemophilic products - by developing incentives. The same should apply to medical devices and diagnostics for rare diseases in general.

**Question 10: What kind of specialised social and educational services for RD patients and their families should be recommended at EU level and at national level?**

The treatment of haemophilia is a good example of what comprehensive care could mean in practice for patients suffering from rare diseases.

Patient organisations have a key role to play in the context of comprehensive care as they can help patients feel empathy and share experiences on the disease. The European Commission should continue to recognise the importance of these patient organisations at the EU level to ensure patients’ empowerment and their increased involvement in the policy making process.

For the treatment of haemophilia, the social and psychological framework is of utmost importance. Social and educational services should be seen as part of the medical treatment (especially for children and teenagers) from the diagnosis to the everyday life with a chronic illness for the patient, their family and carers. The comprehensive care treatment of this complex disease can be delivered in comprehensive care centres which are essential to a better quality of life for patients.

We would like to stress that psychological support should be made available to patients and their families (and care givers) whenever necessary.

*Specific services*

The different services available in these Comprehensive Care Centres can be used as a benchmark for comprehensive care of other rare diseases, as they should include:

- medical services with experienced doctors (e.g. haematologists) and nurses
- laboratories (including genetic ones)
- dental services
- genetic counselling
- physical therapy and physiotherapy services
- psychological support
- community liaison services including home and school visits
- social and vocational services
- home treatment provision

We would also like to stress that patient training (e.g. for home treatment of haemophilia) plays a very important role in patients’ empowerment and in their social integration.
Question 11: What model of governance and of funding scheme would be appropriate for registries, databases and biobanks?

The EHC supports the European dimension of registries and databases, while respecting data protection regulations. Reaching a critical mass when collecting data can allow reaching significant conclusions on the number of patients across Europe, the cause of mortality, etc.

For the particular case of haemophilia, collecting data on epidemiology and morbidity, on causes of mortality and on adverse effects of treatment at the EU level would create strong dynamics and boost national initiatives in the treatment and care of haemophilia patients.

We believe that public funding is a prerequisite for the establishment of registries and database in the field of haemophilia and other Rare Diseases.

The adoption of policies introducing dedicated funding for RD services and networks should be encouraged in the different EU member states.

Research

A real European network for research in the area of rare diseases should also be a priority and the FP7 should allow for this to happen in the very near future.

Question 12: How do you see the role of partners (industry and charities) in an EU action on rare diseases? What model would be the most appropriate?

The EHC believes that the industry and NGOs have a key role to play and joining efforts is the only way forward, especially when dealing with rare diseases.

The EHC is a good example of partnerships with different stakeholders including other patient associations, medical experts and the industry to raise awareness on this disease.

Public/private partnerships with patient organisations, public health authorities and pharmaceutical companies are a good model. Patient organisations are of course a key element of these partnerships as they are the conveyors of messages to their members, the industry and public health authorities with the aim for instance to intensify research efforts and disseminate information amongst patients and other health organisations.

Question 13: Do you agree with the idea of having action plans? If yes should it be at national or regional level in your country?

National action plans for rare diseases are an excellent idea. We know that France has set up a national action plan for rare diseases (2005-2008) and that other countries are also developing strategies and plans. Spain has for instance announced on 23 January 2008 that it would prepare a national action plan on rare diseases, thus following the steps of Romania, Portugal, Italy and Greece.
National plans should be developed according to European guidelines to ensure the coherence and complementary nature of these strategies at the European level. The plans should set very specific objectives and be assessed regularly to ensure that the rhetoric is followed by concrete actions in each member state to improve patients’ treatments and quality of life.

**Question 14: Do you consider it necessary to establish a new European Agency on RD and to launch a feasibility study in 2009?**

EHC would welcome the set up of an EU Agency with the main aim of implementing on a systematic and rigorous basis the RD policies at Community level. It should serve as a core instrument to exchange information, encourage the set up of national action plans and intensify research in the area of rare diseases such as Haemophilia. The set up of this agency should not be a pretext for the Community and national authorities to slow down their efforts to deal with rare diseases but it should be the springboard for a coordinated and harmonised action in this field at the EU level. The feasibility study should start in 2008 to ensure that the Agency is set up early and Community policies start being implemented as soon as possible.

*Implementation reports on the RD communication*

The Commission’s regular implementation reports on the Communication will be an effective way to show the commitment of the Community (and member states) to make progress for better diagnosis, treatment and care of patients suffering from rare diseases. They should of course cover the actions undertaken by the Agency to ensure that this instrument is effective in its activities and really adds value to the implementation of the Community RD policies.

In conclusion, the EHC would like to stress the importance of research (and more particularly genetic therapy) to improve the treatment of rare diseases such as Haemophilia. We know that cross European research into rare diseases such as haemophilia can help finding solutions for other (rare but not only) diseases and further progress should be encouraged and supported by the Commission and its Member States via the FP 7. The concept of comprehensive care, for which the EHC has been advocating for a long time, should also be extended to all rare diseases and should be implemented across the European Union.

**EHC is very pleased that the European Commission has taken the initiative of this Communication and will continue contributing to the debates at the EU level. Most of the questions of the consultation can only receive a positive answer but we hope that this text will be adopted by the Council under the French Presidency in the 2nd half of 2008 and will be followed by concrete action and objective assessment to improve the quality of life of patients suffering from rare diseases, including haemophilia, and their families.**

For further details, please contact EHC at info@ehc.eu; or visit the website www.ehc.eu.
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