The French authorities' response to the European Commission's public consultation on:

"Rare diseases: Europe's challenges"

The French authorities welcome the European Commission's decision to launch a public consultation on "Rare diseases: Europe's challenges".

Given the low prevalence of the 5000 rare diseases identified to date, and hence the limited number of patients suffering from each disease, and given the specific characteristics of these diseases, France has always believed that a pan-European approach to this issue is appropriate. Pan-European cooperation provides very high, essential added value, considering the limited and dispersed nature of research and expertise, and the inadequate information available within each member state.

The proposals put forward emphasise the need to improve knowledge of these diseases, expand research, share knowledge, structure supply of care via setting up Centres of Reference and/or Expertise for the care of rare disease patients at member state level (in accordance with individual national policies) and, at the EU level, create European Reference Networks to address the issue of rare diseases.

In recent years, France has taken several initiatives within the framework of the EU, particularly in the following areas:

- Orphan drugs: France instigated the introduction of Regulation 141/2000 on Orphan Medicinal Products of the European Parliament and of the Council of 16 December 1999, which is widely recognised as being one of the EU's most useful public health policies,
- Information on rare diseases: through the development of the "ORPHANET"[1] database, which has also enjoyed the unerring support of the European Commission,
- Facilitating cooperation between member states, in terms of funding research into rare diseases through the ERA-NET project, "E-Rare" [2].
- "European Reference Networks" for rare diseases: France chairs the Working Group on this subject, deriving from the Commission's High-Level Group on Health Services and Medical Care[3].

[1] See: www.orpha.net/
[2] See: http://www.e-rare.eu/cgi-bin/index.php.
[3] See: http://ec.europa.eu/health/ph_threats/non_com/rare_8_en.htm
See: http://ec.europa.eu/health/ph_overview/co_operation/mobility/high_level_hsmc_en.htm

In France, the public health law of 9 August 2004 identified rare diseases as one of the five national public health priorities, on a par with cancer, chronic diseases, and the impact of violence and environmental factors on health. As a result, a "National Plan for Rare Diseases (2005-2008)"[1] has been developed. The various objectives of this plan are very much in line with the proposals put forward by the Commission in its public consultation document. In particular, one of the ten strategic axes of this national plan is to develop pan-European partnerships to address the issue of rare diseases.

France wishes to ensure that its national policy on rare diseases is consistent with European policy, especially in regard to research, epidemiology, the development of diagnostic tests, European reference networks, patient networks and information in the broad sense of the term.

France intends to pursue its efforts at the European level, especially when it takes over the Presidency of the European Union in the second half of 2008. It is particularly interested in the Commission's work, in view of adopting a recommendation from the Council.

The French response comprises two parts:

Part 1: summary analysis

- the objectives of pan-European cooperation,
- the priority objectives identified by France,
- the organisation and resources needed to implement these objectives.

Part 2: answers to the 14 questions put by the Commission[2]

An appendix on research[3] into rare diseases is attached to this response paper: it reviews all the proposals drawn up by the French Task Force on "Research and Rare Diseases". This Task Force, headed by the Department of Health (under the Ministry of Health, Youth and Sport), was consulted on the drafting of this response paper.

[1] See: http://www.sante.gouv.fr/htm/dossiers/maladies_rares/plan.pdf

[2] ***Please see the answers to the 14 questions as presented (in French) in the official contribution from France***

[3] ***Please also see appendix as presented (in French) in the official contribution from France***

Part 1

Summary Analysis

I- Objectives

As far as France is concerned, the objectives of this pan-European cooperation are as follows:

- develop knowledge of rare diseases by increasing support for research,
- guarantee equal access to diagnosis and appropriate care, by improving information on rare diseases and sharing expertise,
- develop new treatments and make sure that they are available in all member states.

These objectives fully justify the Europe-wide optimisation and sharing of resources that are, by their very nature, limited at member state level. Hence, France is particularly interested in the proposals set forth in the public consultation document, and has identified five priority axes for the development of an EU policy on rare diseases.

II- The five priority axes defined by France for pan-European cooperation on rare diseases

II-1 Improve information on, and knowledge of, rare diseases

This essentially covers the work accomplished by the "ORPHANET" database since 2000. This database is an essential response to the needs and expectations of patients and healthcare workers in charge of rare diseases. France fully supports the activities carried out under the framework of "ORPHANET", on the initiative of its leader Dr. Ségolène Aymé from the INSERM (FR). Furthermore, Dr. Ayme is head of the EU Rare Diseases Task Force, set up in 2004 by DG SANCO. The continued funding and development of projects related to the "ORPHANET" database, through the EU Public Health Programme 2008-2013 and the 7th Framework Research Programme, is decisive in the development and consolidation of European policy on rare diseases. However, other means of communication (besides the Internet) should also be promoted, to ensure equal access to information for patients and the general public alike: telephony, personal patient cards, etc.

France also supports the Commission's proposal to improve the classification of rare diseases. This would allow for reliable and uniform epidemiological monitoring of rare diseases throughout all the member states. This knowledge tool is essential to individualising rare diseases within information systems, and is necessary to defining public health priorities (cf. response to question 2).

II-2 Strengthen research into rare diseases

France wishes to reinforce the Commission's proposals to accelerate research into rare diseases. Rare diseases are a particularly sensitive issue, in which the transfer of research findings to the clinical setting is instrumental to the development of innovative therapies within the shortest possible timeframe. The level of European collaboration is particularly decisive in the research sector, at all stages of the process ***(cf. appendix on Research).***

II-3 Set up and develop European Reference Networks for rare diseases

At present, there are several different types of European network, such as thematic networks of expertise and/or reference centres dedicated to patient care, networks of registers and research and/or information networks.

These networks must be brought together and harmonised, as they are valuable tools in the implementation of EU policy on rare diseases, in close collaboration with patients' organisations. They should be given priority support on a permanent basis, provided that implementation and evaluation criteria are met. These European Reference Networks should privilege the transfer of expertise, rather than the mobility of patients.

Hence France supports the proposals developed since 2004 by the working group on "European Reference Networks". This Working Group (which is chaired by France) derived from the DG SANCO's High-Level Group on Health Services and Medical Care. It defined the concept of reference networks, as well as the criteria and specifications that they must meet and the practical procedures governing their implementation. In 2006, the EU Public Health Programme funded a number of pilot projects.

The development of European Reference Networks, as specified by the High-Level Group, should be based on the development of medical information sharing tools (electronic medical records, exchange of data on biological samples, imaging, etc.). These networks are valuable tools that should, for the benefit of patients and healthcare workers alike, be expanded to play a central part in EU policy on rare diseases.

The initial and continuing education of healthcare workers, and the sharing of knowledge, are also of prime importance in improving the treatment of rare disease patients. The European Reference Networks could make a useful contribution in these areas.

II-4 Pool expertise on rare diseases

The objective here is to set up a system for summarising and sharing the scientific information available across Europe, so that individual member states can draw on it to make decisions in different areas, such as prevention, screening, diagnosis, treatment and the post-marketing authorisation evaluation of medicinal products (in terms of the medical service rendered).

The pooling of information should ensure that scientific progress in all these areas is available to as many people as possible, and should be used as a basis for drafting a set of European best practice recommendations, similar to the guidelines on cancer screening drawn up since 2003. Each individual member state would remain responsible for the implementation decision and procedure, in accordance with the principle of subsidiarity.

Finally, France would like the results of all projects on rare diseases, financed by the European Commission (DG SANCO, DG Research), to be widely published and promoted.

II-5 Support international cooperation on rare diseases

In addition to European cooperation, France suggests that the future Communication on rare diseases should provide an opportunity for more large-scale cooperation, i.e. on an international level:

- between developed countries, in particular the United States, Canada, Japan, Singapore, Korea and Australia. Indeed, international research policies relating to rare diseases and orphan drugs are also necessary and relevant. It would also enable patients suffering from very rare, or even extremely rare, diseases to be grouped together;

- between North and South, as well as with Eastern European countries outwith the European Union, i.e. countries in the Mediterranean basin, Africa, the Caribbean and Pacific with close ties to EU member states. This would encompass, in particular, access to information, the sharing of treatment expertise, and the development of healthcare worker and patient networks.

III- The organisation and resources needed to meet these objectives

The implementation of these five strategic axes, with a view to enhancing EU cooperation on rare diseases, will only be possible if a permanent organisational structure is set up, benefiting from permanent resources.

III-1 Organising European cooperation

France supports the Commission's proposal to create a "European Advisory Committee on Rare Diseases", the aim being to provide a permanent organisational structure and permanent funding for the very specific area of rare diseases. France is in favour of this type of flexible, permanent organisation and would like this Committee to be cross-sectoral and to include:

- representatives of the health authorities from the different member states;
- representatives from the Commission's main Directorate Generals, concerned by this issue (e.g.: Health and Consumer Protection; Enterprise and Industry; Science and Research; Employment, Social Affairs and Equal Opportunities; Internal Market; Information Society; Education and Culture; Development) ;
- recognised experts in the field of rare diseases,
- representatives of patients' organisations, who are key players in European policy in this field.

France would like the scope of this Committee to include the definition of the strategic objectives of European policy on rare diseases, the coordination of actions, monitoring and annual evaluation. One of its tasks should also be to lead sustained discussions with both

academic research bodies and companies concerned by the different aspects of research into rare diseases (medicines, diagnostic tests, etc.).

France is opposed to the creation of a “European Agency on Rare Diseases", insofar as the objectives of such an agency would be the same as those of the Advisory Committee. France is therefore against the idea of launching a feasibility study on the creation of a new agency on rare diseases (cf. response to question 14).

III-2 Resources

France is strongly in favour of allocating a specific, permanent budget for the running and activities of the Advisory Committee, which should therefore appear as such in the EU budget over the coming years.

In addition to providing funding for the Advisory Committee, and in order to guarantee the medium- or even long-term implementation of this European policy, France is in favour of two things:

- firstly, the principle of providing renewed financial support for calls for proposals and tenders in the field of academic research, and for research into rare diseases conducted under the framework of public/private-sector partnerships,
- secondly, permanent funding for particularly structure-building activities.

This includes:

- support for the “ORPHANET” database and the development of information on rare diseases;
- support for rare disease patients’ organisations, which are a source of proposals and opinions regarding European policy on rare diseases;
- the financing of “European Reference Networks”, which are essential tools to reinforcing dialogue and cooperation between the member states, in different areas of research, epidemiology, care, knowledge sharing, training, etc.
- the financing of bio-banks, registers and databases, which should be accessible to research teams throughout Europe.

Concerning the **sources of funds**, France is in favour of:

- identifying and setting up a specific EU fund for rare diseases, as proposed by the Commission;
- keeping on treating rare diseases as a priority in EU public health programmes and research;
- developing public/private-sector partnerships, under the express condition that independence rules are observed, especially in regard to healthcare workers and patient rights, in the areas of research, technology transfer and the compassionate use of medicinal products.

Finally, France supports the last proposal in the document, relative to the production of an annual report on the situation of rare diseases in the European Union. This report will be used to assess the implementation of the future Communication on rare diseases in the different member states, and to tailor any required measures. It is also in favour of organising - in tandem with the European Advisory Committee - European conferences involving all stakeholders. These conferences could be held bi-annually, as has been the custom since 2001.

This paper represents the views of its author on the subject. These views have not been adopted or in any way approved by the Commission and should not be relied upon as a statement of the Commission's or Health & Consumer Protection DG's views. The European Commission does not guarantee the accuracy of the data included in this paper, nor does it accept responsibility for any use made thereof.