European Commission
Health and Consumer Protection Directorate-General
Rare Diseases Consultation
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[already in English]

Attachment:
Consultation statement, public consultation.doc

Consultation statement

Rare diseases – Europe's challenges

In a public consultation from "The Health and Consumer Protection Directorate-General" in the European Commission on *"Rare diseases – Europe's challenges"*, input is requested before 14 February 2008. This input is from the Norwegian Directorate for Health and Social Affairs (*Shdir)*. The document is written in Norwegian with the Danish version of the consultation as a basis.

The questions are commented on in chronological order:

1. Improving the identification and knowledge of rare diseases

*** Common definition of rare diseases in the EU:** The existing definition of rare diseases is established in the Community's 1999 – 2003 rare diseases action programme as diseases that present a prevalence of **fewer than 5 in 10 000** people in the EU. The EMEA uses the same definition to identify medicines for rare diseases (regulation) and several Member States which have taken specific initiatives (France, Germany, Italy, the Netherlands and Spain). The United Kingdom, Sweden and Denmark use other definitions, however. Even if some partners perceive the current definition as being too broad, the EU would prefer to keep the current definition.

Question 1: Is the EU's current definition of a rare disease satisfactory?

Comment: In Norway, a rare medical condition is defined as a condition where there are fewer than 100 known individuals per million inhabitants in the country. This is based on a cooperation project concerning small and less well-known disabled groups under the Nordic Council of Ministers from the early 1980s. Denmark has since adapted its definition to the EU. There should be discussion as to whether or not the other Nordic countries should also redefine what is considered to be a "rare medical condition".

*** Better codification and classification of a rare disease:** The EU should work closely with WHO to revise the existing ICD (international overview of diseases) to ensure that rare diseases are correctly coded so that they can be traced in all health information systems. This requires support from a working group on the classification and codification of rare diseases that will function as a consultation group for WHO in the ICD revision process. There will also be a need for active cooperation within the EU's statistics programme as soon as the new ICD-11 exists, in order to ensure application of this version and the use the new codes for rare diseases in death certificates and hospitals' printout systems in all Member States. There should also be a similar input to ensure that rare diseases are codified correctly in the SnowMed and MedDRA codification systems.

Question 2: Do you agree that there is an urgent need to improve codification and classification in this area?

Comment: There is an urgent need to improve coding and classification of rare conditions in the ICD coding. Today, this is not really suitable for the classification of rare conditions.

*** Creation of a list of rare diseases:** One of the reasons why rare diseases are overlooked is that there is no general knowledge of which diseases these are. It is important to give society a regularly updated and accurate list of rare diseases that are also classified according to special field, prevalence, mechanism and aetiology to increase knowledge and ensure documented support for research and data storage in general. The European Commission ought to offer financial support for this activity under the public health programme.

Question 3: Can a European list of rare diseases help your national/regional systems to improve their handling of rare diseases?

Comment: This requires a common definition of rare diseases. As complete as possible an overview of the diseases that come under the designation of "rare medical condition" according to a common definition will be able to help to improve the handling of rare diseases.

2. Improving prevention, diagnosis and treatment of patients with rare diseases

*** Communication of relevant information:** In order to improve the diagnosis and treatment of rare diseases, it is important to have access to accurate information in a format that is adapted to the needs of the health sector and the relevant persons. Since the year 2000, it has been possible to obtain information on over 5 000 rare diseases in six languages in the **Orphanet** database. This contains a comprehensive encyclopaedia of rare diseases, a list of health services in 35 countries, a list of European reference centres, a database of medicines for rare diseases with information on the development stage and availability in the EU countries, plus a number of other services for specific partnership groups, including a facility for obtaining diagnoses with the help of symptoms and signs, and finally a library of recommendations in emergency situations. The European Commission ought to offer more financial support for this activity under the public health programme and FP7.

*** Support for information networks:** Here, the priority is on the exchange of information through existing European information networks to achieve better classification, develop strategies and mechanisms for the exchange of information between partners, define relevant health indicators, develop comparable epidemiological data at EU level, support the exchange of good practice and develop measures for patient groups. Ongoing projects have already shown their relevance. Support for this form of project ought to continue in both the EU and the Member States. Support for specific international consensus conferences also appears to be relevant. The European Commission ought to offer financial support for this activity under the public health programme and FP7.

*** Development of national/regional reference centres and the setting up of reference networks at EU level:** When a disease is rare, there is also not much expert knowledge on it. Some expert centres (also called reference centres) have developed an expert knowledge that is used widely by other specialists in their countries and possibly also internationally. These centres are publicly recognised in some countries, but in most countries they function exclusively on their reputation. The Commission has decided that the most effective access will be to prioritise cooperation and knowledge distribution between these centres. Certain principles have been developed for European networks (ERN), among other things for their role in the handling of rare diseases and other diseases that require special treatment, and for patient numbers and some other criteria to be fulfilled by these centres. ERN must also function as a research and knowledge network that updates and contributes to the latest scientific results, treats patients from other Member States and ensures subsequent access to treatment facilities if this is necessary. The definition of ERN must also reflect the necessity of an equal distribution of services and expert knowledge in the enlarged

Union. In 2006, the EU's rare diseases task force drew up a report entitled *'Contribution to policy shaping: For a European collaboration on health services and medical care in the field of RD'*[22], where it suggests that the Member States make a contribution towards recognising their expert centres and offer them as much financial support as possible. It also recommends that the Member States draw up guidelines for their patients to help them find their way through the health service; this will take place through the establishment of cooperation between all necessary expert centres in the country, and also abroad if necessary. It also suggests that financial support should continue to be offered for reference networks of expert centres under the relevant EU programmes within rare diseases until there is an evaluation of the result of the network processes.

Question 4: Should the European reference networks prioritise transfer of knowledge, mobility of patients, both, how?

Comment: 'Sjeldenfeltet' [the rare case] in Norway is based on Competence Centres for rare and little-known diagnoses and function inhibitions, as well as the ordinary health and social services. The transfer of knowledge between different competence centres within Sjeldenfeltet ought to be a priority, particularly concerning the rarest of conditions. In Norway, patient mobility where treatment is concerned is regulated by the offices for medical treatment abroad in each regional health authority. You can apply for support for courses/sessions abroad three times for the same child up until the age of 18. Norway therefore already has an existing arrangement in this field.

It will be important to involve users in this in all cases. User participation ought to be included as a natural part of any knowledge build-up and communication.

*** Development of e-health within rare diseases:** Electronic services developed by Orphanet and other EU-financed projects clearly show how e-technology can be involved in putting patients in contact with one another, that databases are shared between research groups, collecting data for clinical research, registering patients who are willing to participate in clinical research, and forwarding records to experts who can improve the quality of the diagnostics and treatment. Online tools and electronic tools are very efficient and ought to form a strong part of the EU's strategy for rare diseases. They can save the life of a person with a rare disease in a crisis situation. The European Commission ought to offer financial support for this activity the public health programme and FP, as well as through the Member States.

Question 5: Should online tools and electronic tools be developed in this area?

Comment: We recommend the continuation/development of already existing tools such as www.orpha.net. In the Nordic Countries, there is more investment in the development of the collection of links, www.rarelink.no.

*** Availability of and access to accurate diagnostic tests, including genetic tests:** Many rare diseases can now be diagnosed using a biological test that is often a genetic test. These tests are important elements of good patient management, since they facilitate diagnosis at an early stage and often make it possible to undertake cascade screening of a family or a prenatal screening. Due to the large numbers of tests and the need to develop and validate a specific set of diagnostic analyses for everyone, each individual country cannot be self-sufficient in this field. Patient records and tests are therefore exchanged across national borders. This flow over the borders is quite clearly a mechanism that can fill a major gap in the availability of tests for rare diseases. This exchange ought to be promoted through clearly established, transparent standards and procedures that are established at EU level. It is necessary to compensate for the legislative differences between the countries as regards practice concerning confidentiality, remuneration, transport of tests and storage and certification of laboratories. The laboratories ought to be encouraged to participate in **performance tests** with particular emphasis on reporting results. Genetic consultation ought to be developed concerning pre and post testing. This requires support at the relevant level (depending on the number of tests per year) for reference laboratories. Over the past two years, the various partners (European Commission[23], The Council of Europe and particularly OECD[24]) have made a significant contribution to introduce a quality assurance policy for laboratories.

Question 6: What can be done to further improve the access to quality tests for rare diseases?

Comment: In Norway, a database of a range of tests has been established at the country's medical genetic laboratories (comes under www.medgen.no). The overview in Orphanet can be further developed. There are also other overviews of the range of tests, such as http://www.ukgtn.nhs.uk/gtn/ and http://www.eurogentest.org/. The latter may be fortified as an official EU database.

*** Evaluation of strategies for the screening of populations (including neonatal screening) for rare diseases:** Neonatal screening for PKU and congenital hypothyroidism is general practice in Europe and has proven to be extremely effective with regard to preventing disability in the relevant children. In line with the technological development, many tests can now be performed at low cost (including robotic tests) for a large number of different rare diseases, particularly metabolic disturbances and genetic diseases in general. However, this ought not to constitute sufficient grounds to introduce them into the policies for population screening without first doing a careful evaluation in relation to WHO's criteria from 1965 (to be checked), since screening can be harmful to those who are screened and uses a great many public resources. There is currently only a limited consensus as to which diseases benefit from a systematic screening in accordance with the WHO criteria. The implementation of targeted screenings or population screenings depends on many things such as the quality and reliability of the tests, access to effective treatment/surgery for those who are screened, the prevalence and seriousness of the disease as well as the value that society attaches to screening. It is recommended that cooperation be encouraged in the area to bring about documentation on which the Member States can base their decisions.

Question 7: Do you think that it is important to evaluate potential population screening for rare diseases at EU level?

Comment: The overall understanding ought to be followed here, rather than establishing screening tests just because it is technically possible. The health benefit must be clarified. Cf. Book: Holland, WW, Stewart, S.: "Screening in Disease Prevention. What works?" The Nuffield Trust, Oxford 2005.

User participation ought to be ensured in the drawing up of any new screening service.

*** Primary preventative measures when possible:** It is possible to undertake primary prevention only for very few rare diseases. Environmental factors are important elements of a broad range of rare congenital abnormalities and cancer in children. In order to prevent these rare diseases, public health measures concerning health determinants must be undertaken specifically during the period before fertilisation and during pregnancy – nutrition, obesity, alcohol, smoking, use of recreational substances and environmental contamination. If people are vaccinated against diseases like Rubella (to prevent congenital rubella syndrome), migration between countries with different vaccination policies ought to be taken into account. In the management of chronic diseases such as diabetes, epilepsy and infertility, emphasis ought also to be placed on women before fertilisation and at the start of the pregnancy. Possible treatments include a greater intake of folic acid for women before fertilisation to prevent spinal cord abnormalities (e.g. spina bifida) and other abnormalities in the foetus. Many examinations show that the periconceptional ingestion of correct amounts of folic acid during (sic) can prevent more than half of spinal defects. Initiatives in this field ought to be discussed at EU level so that it can be established for which rare diseases preventative measures can successfully be introduced.

***Good practice for the treatment of rare diseases:** The identification and description of good practice is crucial to the exchange of information and data on effective strategies for the treatment of rare diseases and therefore also to improve the level of information and knowledge of good practice for the treatment of rare diseases. The exchange of good practice makes it possible for the EU's Member States to utilise other people's experiences and thereby build up networks between different treatment services that work with rare diseases. Benchmarking at Member State level will increase the chances of successful treatment of rare diseases.

***Equal access to medicines for rare diseases:** In spite of good incentives to develop and register medicines for rare diseases, citizens' access to treatment that can save their lives is limited by two factors. Firstly, not all businesses offer their market-approved products in all Member States, which is due to registration problems at Member State level. Secondly, there have been examples of administrative delays (way beyond the maximum of 180 days) in giving the public access to the approved medicines. This leads to substantial differences in the number of available medicines in the Member States. This situation ought to be changed. The Commission ought to produce a report for the Council and the Parliament every other year with a description of these bottlenecks (delays, marketing, access, remuneration, prices, etc.) with proposals for the necessary changes to the law that will ensure equal access to medicines for rare diseases throughout the EU. The hospitals' medicines for rare diseases will be financed at a higher administrative level than the local hospital to ensure capacity to supply these medicines to patients.

Question 8: Do you expect the problem with access to medicines for rare diseases to be solved at national or EU level?

Comment: SHdir chooses not to say anything on this point.

*** Medical equipment for and diagnosis of rare diseases:** The Regulation on medicines for rare diseases does not cover the field of medical equipment and diagnostics. The limited size of the market does prevent the industry from developing products for patients suffering from rare diseases, however. Initiatives for the development of incentives for the medical equipment and diagnostics industry ought to be sought on the basis of this model that has been used for medicines for rare diseases.

Question 9: Should the EU have a Regulation on medical equipment and diagnostics?

Comment: SHdir chooses not to say anything on this point.

*** Health technology assessment of medicines for rare diseases:** Health technology assessments for rare diseases that are to be undertaken before prices and remuneration are established constitute another factor that is beginning to play a crucial role in patients' late access to the medicine, or that even prevents them from being treated. The methods that are used to evaluate the cost-effectiveness of medicines on the general market do not apply to medicines for rare diseases. It is usually not possible to make comparisons, and there are only limited quantities of data. Ethical access to the subject also cannot be based solely on economic criteria, since the economic evaluation constitutes just one of the elements in the decision process where consideration ought to be given to society's preferences. There is a need for coordinated access to this subject in the Member States. Research also ought to be encouraged in relevant evaluation methods that are based on the patient perspective.

***Coordinated special delivery programme:** There is a need for a better system to deliver medicine to patients before approval and/or indemnification (compassionate use, special delivery) of medicines. The responsibility for special delivery of medicines ought to lie jointly with the doctor, the product developer and the authorities. It ought not to be forgotten that a number of medicines for rare diseases are developed by small and medium-sized companies that cannot afford to follow special delivery programmes without public support. This question ought to be coordinated between the Member States with support from the European Commission. Article 83 in Regulation (EC) no. 726/2004 gives the Member States the option of undertaking special deliveries, and EMEA can adopt an opinion on the terms for using a medicine for rare diseases if there is a question of special delivery.

*** Special social services** are important to improve the quality of life for people with a rare disease. These social services include the following that have been identified as particularly well suited to the improvement of the quality of life for both the patient and those who are looking after the person concerned, who are often family members. **Relief services** give both carers and patients the option of adapting their lives so that they can have breaks with time to themselves. **Information services and help lines** increase patients' and carers' chances of obtaining relevant information on the rare disease with which they are living on a daily basis. **Recreational programmes for**

children and young people give the patients the opportunity to gain a different perspective on life from the one of being ill. **Financial support** is of help to carers who must try to take care of paid work and unpaid care. **Psychological support.** The European Commission ought to offer financial support for psychological assistance under the public health programme and the action plans for the disabled.

Question 10: Which forms of specialised social and training services for patients with rare diseases and their families ought to be offered at EU level and national level respectively?

Comment: SHdir chooses not to say anything on this point.

4.3 Accelerating research and development in rare diseases and medicines for rare diseases

*** Databases, registers, archives and biobanks:** Registers and databases are important instruments in the development of the clinical research into rare diseases. They constitute the only way of collating data and achieving sufficiently large samples for epidemiological and/or clinical research. Registers of patients who are treated with medicines for rare diseases are particularly relevant because they make it possible to obtain documentation on the effectiveness and any side effects of the treatment, since marketing licences are normally given at a time when there is still limited, if convincing, documentation. Support ought to be offered for cooperation regarding the set-up and updating of databanks, provided that this support is covered by fixed rules. Many networks for researchers and health personnel for which support is offered by GD RTD and GD SANCO have set up such common infrastructures that have proven to be very effective tools for increasing knowledge and implementing clinical trials. A special network such as EuroBioBank[26] is an evaluation of the European resource that requires long-term financing and EU-based access if it is to be developed and used optimally. Support ought to be offered for this form of initiative from both Member States and the EU, and long-term support should be allocated to these infrastructures, provided that their beneficial value has been ascertained. The same applies to databanks of biological samples and biobanks. In connection with biobanks for rare diseases, there is a particular need to collate and store documentation on patients with very rare diseases, even if there is no current research protocol. The Member States and the European Commission have offered support for the following areas: quality standards, including the development of strategies and work tools for periodic monitoring of the quality of databases and the cleaning of databases, a common minimum set of data for research into epidemiology and the people's health, focus on user friendliness, transparency and connectivity for databases, intellectual property rights, communication between databases and registers (genetic, more generic diagnostics, clinical, those controlling monitoring, etc.). Emphasis should be placed on creating a link between international (European) and national and/or regional databases if these exist.

Question 11: Which governance and financing model will be suitable for registers, databases and biobanks?

Comment: SHdir chooses not to say anything on this point.

*** Biomarkers:** Biological markers (biomarkers) are "objectively measurable indicators of biological processes". They can be used to diagnose the disease and evaluate its course and reaction to treatment. Many current diagnostic tests (tumour markers, parts of DNA sequences that cause or

are associated with a disease) come under the definition of biomarkers. Functional and radiological evaluations can also be seen as biomarkers. In connection with the evaluation of the course of a disease and potential new treatments, biomarkers can be used instead of natural end points such as survival or irreversible morbidity, end points that require long-term observation and broad patient populations. This applies largely to rare diseases due to the low number of people who are affected by each disease. There are examples of marketing licences having been given on the basis of biomarkers as end points in connection with the assessment of the effectiveness of that agent. The new molecular biological techniques have been the driving force behind research into biomarkers (e.g. genomics, proteomics and combination chemistry) that can be used to identify large quantities of potential biomarkers simultaneously. It is important for the EU to offer support for new techniques for the development of biomarkers, also including radiodiagnostic and functional techniques. However, it is even more important to support investigations and activities aimed at validating potential biomarkers and bringing them into clinical use. This process is long, costly and at this moment ineffective. Within rare diseases, it will be advantageous if support is offered for the evaluation of the validity of specific biomarkers (or groups of biomarkers) on as many patients as possible (reference network), and if the partnership between the pharmaceuticals industry and the academic world is strengthened with a view to translational research, i.e. selling the research results, the patients will benefit.

*** Data protection:** All these infrastructures must be set up in accordance with the EU's Regulations and agreements concerning data confidentiality and protection of the patients' private lives. Emphasis should namely be on **the data protection directive**[27]. The IDA initiative (data exchange between administrations) ought to be implemented, since this will make it easier to establish European registers on specific rare diseases of great relevance to the public.

*** Researcher network for rare diseases:** Coordinated research projects at EU level are crucial to success. Support ought to be offered for coordinated networks from both the Member States and the EU, and rare diseases ought to continue to be strongly prioritised in GD RTD's future programmes. Different new areas such as social research into rare diseases should also be included.

***Coordination between the Member States' financing agencies:** The EU's ERA-NET project, for which support is offered under FP6, and that currently coordinates seven countries' financing policies for rare diseases, is an example of a successful solution to the fragmented research input. This strategy ought to be furthered and more Member States called upon to join this initiative.

***Intensification of the research:** For the majority of serious rare diseases that are potentially treatable there is simply no specific treatment. The development of forms of treatment faces three obstacles: lack of understanding of the underlying pathophysiological mechanisms, lack of public support in the initial phases of the clinical development, and lack of interest on the part of the pharmaceuticals industry. The substantial costs of developing medicines, along with the estimated low investment yield (attributed to the very small patient populations), have prevented the pharmaceuticals industry from developing medicines for rare diseases, in spite of the enormous need. Even if the legislation on medicines for rare diseases has definitely promoted the treatment thereof, there are still major problems and needs for further initiatives. Since the identification of targets for treatment largely depends on the genetic and molecular description of the diseases and the identification of the biological mechanisms, it is of crucial importance to intensify the pathophysiological and clinical research into rare diseases. With the research-related progress, the sequencing of the human genome and the development of high-capacity work tools within genomics and postgenomics, we can expect the mechanisms behind many rare genetic diseases to be identified within the next few years. Support ought to be offered for research into forms of

treatment, including innovative biotechnological research (monoclonal antibodies, cell and gene treatment and enzyme replacement therapy) and more classical treatment research based on research into active chemical compounds. Even within rare diseases, the selection of chemical compounds that react to identified biological targets is an important research objective in medicines. Since the pharmaceuticals industry does not usually wish to carry out this introductory work, it is important to develop an interest in this within the public sector. The EU ought to offer support for academic research into preclinical development. Connection to the European high capacity platforms that are now under construction should also be encouraged, as should the use of common European libraries of molecules. Investigations carried out into the interface between pharmaceuticals companies and organisations in the public sector ought to be supported by the setting up of public private partnerships that will evaluate the medicines applicants. At European level, this challenge can be taken up by establishing and financing a public private forum for rare diseases that will facilitate the development of promising preclinical and clinical projects with the participation of several centres, since this will create the necessary expert knowledge and financing. At Member State level, support should be offered for academic clinical trials on the basis of the model chosen by Italy, France and Spain, and this input ought to be coordinated to ensure the participation of a sufficiently large number of patients.

Question 12: How do you perceive the partners' (the industry's and charitable institutions') role in an EU initiative concerning rare diseases? Which model would be the most suitable?

Comment: SHdir chooses not to say anything on this point.

4.4 Raising the awareness of patients with rare diseases on an individual and a collective level
*** Common strategy for the involvement of patient organisations:** Patient organisations have proven in both the Member States and the EU to be valuable partners who help to make visible rare diseases, collate and communicate background information for a public policy on rare diseases, improve the access to information on and medicines for rare diseases, hold workshops at European and national level and draw up guidelines and pedagogic documents. Raising the collective awareness and greater participation of patients and patient organisations will require support for activities such as capacity building, training and network activities between patient groups at regional, national and European level, the exchange of information, experiences and good practice concerning patient services, as well as the establishment of a "patient support society" for isolated patients suffering from very rare diseases and their families. Support for this should be offered under the public health programme and FP7.

Coordinating policies and initiatives at Member State level and EU level
*** Adoption of national/regional plans for rare diseases:** In order to integrate all of the necessary initiatives that ought to be taken at national and/or regional level, the Member States are called upon to draw up national or regional action plans for rare diseases. Only a limited number of Member States have adopted or are adopting a national action plan or are effecting relevant initiatives. France is the only country that has worked out a comprehensive action plan (2005-2008)[29], while other Member States have introduced national policies in individual areas (Italy, Sweden, Denmark and the United Kingdom) or are in the process of introducing such policies (Bulgaria, Portugal, Spain, Romania and Luxembourg). Other Member States have only a targeted policy on the research area (Germany and the Netherlands). The EU strongly encourages the

adoption of national/regional action plans in accordance with the recommendation in the communication, and the subsequent coordination of these. European guidelines on the drawing up of action plans for rare diseases may be useful. This will support the EU's policy of "ensuring equal access to health services and controlling their price and quality". This support is one of the priorities of the public health programme.

Question 13: Do you agree that action plans are a good idea? If so, should they be at national or regional level in your country?

Comment: The drawing up of action plans for the work for people with rare conditions in each country may be beneficial. Such plans ought to be drawn up at national level in order to achieve a uniform approximation and an equal service to all of the country's citizens with rare conditions. User participation ought to be ensured when drawing up action plans.

*** Development of health indicators for rare diseases:** It is necessary to develop health indicators to be able to consider the current and future number of people affected by disease in the EU. The collation of existing data sources should be encouraged, particularly sources that are financed at EU level. A set of realistic and relevant indicators ought to be established for the availability of and access to medicines for rare diseases, for expert/reference centres and for policies at Member State and EU level.

*** A European conference on rare diseases:** Since 2001, European conferences have been held on rare diseases every other year (Copenhagen 2001, Paris 2003, Luxembourg 2005 and Lisbon 2007[30]). They have been invaluable since the participants have been able to exchange information and experiences and they have functioned as an effective communication instrument that has been used to ensure that rare diseases are made visible in the media. They should function as a platform for patients, health sectors and politicians who gain access to information on policies, strategies and examples of successful initiatives; here, they can be put in order and recount their requirements, promote patient-centred policies at national and European level and confirm that the society for rare diseases in Europe is vital and living. The conference ought to be held in cooperation with the EU's advisory committee on rare diseases.

***Setting up of the EU's advisory committee on rare diseases:** The EU's advisory committee on rare diseases will carry out the tasks that have thus far been taken care of by the EU's rare diseases task force. The committee is supported by a scientific secretariat that will help to develop people's health initiatives within rare diseases and be competent to advice the Commission on: (i) the establishment of services concerning rare diseases based on the national action plans (subsidiarity), (ii) clinical tests and screening, (iii) approval of reference networks for rare diseases and quality control, (iv) development of guidelines for good practice, (v) the periodic epidemiological report on the situation of rare diseases in the EU, (vi) registers/networks/ad-hoc investigations at EU level, (viii) support for the development of policies at EU level, (viii) the establishment of common frameworks for the people's health in the area, (ix) the drawing up of an electronic newsletter on rare diseases. The EU's advisory committee on rare diseases will consist of representatives of the most relevant patient organisations and of the Member States' health authorities. The committee will emphasise ongoing and previous projects on rare diseases for which support was offered under the public health programme, but also focus on a broad section of projects on rare diseases under FP. In order to ensure freedom of action for the committee, a specific budget ought to be allocated in the EU's budget for the forthcoming years.

*** Rare diseases in the EU's budget:** All initiatives that are financed by the European Commission are financed on the basis of short-term contracts. Even where there is a regular evaluation of the efficiency of the projects and their relevance in relation to the EU's policy, it is difficult and sometimes impossible within the current frameworks to extend them, which is seen as a serious obstacle to the development of common infrastructures. Another cornerstone in the EU's forthcoming public health programme (2014-2020) for rare diseases should be the establishment of a fund for rare diseases, information services, genetic accreditation and accreditation of laboratories concerning rare diseases, the sustainability of the European knowledge platform for registers and databases concerning rare diseases, as well as other activity in the area for which there is a need for sustainability and long-term financing, which will be described in the implementation reports on this communication from the Commission to the Council and the European Parliament.
*** Setting up of a Community Agency for rare diseases:** A European agency ought to be set up to ensure the long-term implementation of the policies on rare diseases at Community level. An agency is defined as follows: "*A Community agency is a Community legal body. It differs from the EU institutions and has the status of a legal person. It is set up through a procedural document that comes under the secondary EU Law with reference to solving a* ***specific technical, scientific or management assignment***". An EU rare diseases agency may be an excellent instrument to ensure continuity and context in the relevant strategies at EU level in different areas such as patient registers, biobanks, clinical trials, information on rare diseases, network of reference centres, the establishment of common recommendations for clinical treatment and quality assessment. On the basis of GD SANCO's work and advice from the European advisory committee on rare diseases, the European Commission ought in 2009 to initiate an implementation study concerning the setting up of a European rare diseases agency. The agency ought to be the cornerstone in the forthcoming EU public health programme (2014-2020) in the field of rare diseases.

Question 14: Do you think it is necessary to set up a new European rare diseases agency and initiate a viability study in 2009?

Comment: We consider it to be superfluous to establish a new European agency in this field. Resources also ought not to be used on specialist cooperation between the nations. It does not appear to be appropriate to have a new bureaucratic section at EU level.

***Regular reporting on the situation of rare diseases in the EU:** The Commission ought to draw up an **implementation report** every three years on the communication to the Council, the Parliament,

the Social and Economic Committee and the Regional Committee containing a description of the situation and the epidemiology of rare diseases in the EU, plus on the implementation of the Commission’s communication on rare diseases.