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Re: Public Consultation – Rare Diseases: Europe’s Challenges

Dear Mr. Montserrat,

Thank you for the opportunity to comment on the European Commission consultation paper “Rare Diseases: Europe’s Challenges”.

EuropaBio and EBE’s joint Taskforce on Orphan Medicinal Products welcomes the initiative of the European Commission to conduct a public consultation on rare diseases. Patients with rare diseases have the same right to treatment and prevention as any other patients. The combined focus in the consultation paper on research, development and market access for Orphan Medicinal Products (OMPs) is also welcomed – as it is the latter that is creating the current bottleneck.

The spirit of the EU Orphan Medicinal Product Regulation has always been to provide timely and equitable access to therapies for patients with rare diseases and to balance risk by providing economic and other incentives to industry to develop such therapies. Patients with rare disorders deserve the same care and the same safety, efficacy and quality of products as patients with more common diseases. But, while access to new orphan medicinal products is discussed in the consultation paper, the companies involved in the orphan drugs field believe that more emphasis should be placed on why access to existing treatments for rare diseases is a problem. Implementation of the OMP Regulation can only be truly successful if patients who need the products have access to them.

Such improvements will come faster with better communication of the economic and social value of orphan drugs. As well as addressing individual patient needs, we believe that a wider societal perspective must be applied when making decisions that determine the value of orphan drugs. There are direct societal benefits such as allowing patients and carers to enter the workforce. However, there are also indirect societal benefits such as the generation of knowledge of new mechanisms which could lead to the development of new treatments for more common diseases. Increasing awareness of this point is essential to improving access to these treatments. The whole notion of ‘value’ and ‘value for whom’ needs to be examined, as do communication tools to create awareness about the ramifications of removing access to these treatments.

Our key messages are simple:

- Greater acknowledgment amongst decision-makers that an orphan medicinal product is a product to treat life-threatening or serious debilitating conditions for which no, or no satisfactory, alternative exists in the EU, or if such methods exists, the product would be of significant benefit to those patients affected by the condition.
- Creative solutions are needed to address the challenges raised by rarity, at all stages of the process. The challenges facing the research, development of products for treating rare diseases are a direct result of the rarity and severity of the disease in question. The rarity of these conditions makes every stage of the process more challenging – from clinical development, through authorisation, up to and including reimbursement decisions. These challenges increase in direct correlation to the increasing rarity.
- More awareness about the impact of these rare diseases on national healthcare systems is needed. The way to undertake more research (including on understanding risk) is to build partnerships with other stakeholders.
- There is a need to collect data about all rare disease initiatives and orphan medicinal products in Europe and to share data on outcomes and best practice in diagnosis, treatment and care.
- Industry is willing to work in partnership with the healthcare systems to make life-saving treatments available to patients in a timely manner and to support the development of sustainable systems that can deliver expert care to patients with rare diseases.

The ideal is to create a sustainable healthcare system capable of caring for rare disease patients, while enabling companies to have a business model allowing a return on investment and, therefore, continue their work in the rare disease field. The components of the system would include educational programmes to ensure a high level of awareness among patients and healthcare providers, expert centres where patients can be evaluated by knowledgeable care-givers with access to appropriate testing, both diagnostic and for ongoing evaluation, and ultimately access to the best therapies.

Thank you again for the opportunity to contribute. We look forward to being involved in the next stages of this process and to ensuring that innovative treatments for rare diseases reach those Europeans who need them.

Yours sincerely,

Dr. Erik Tambuyzer
Chairman
EBE/EuropaBio Task Force on Orphan Medicinal Products

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EuropaBio

EuropaBio is the European Association for Bioindustries, solely and uniquely bringing together bioscience companies from all fields of research and development, testing, manufacturing and distribution of biotechnology products. It has 84 corporate members operating worldwide, 8 associate members, 6 BioRegions and 25 national biotechnology associations representing some 1800 small and medium sized enterprises involved in research.

EBE

EBE (European Biopharmaceutical Enterprises) is the European trade association that represents biopharmaceutical companies of all sizes operating in Europe. It has 65 member companies, which are engaged in research, development and marketing of new medicinal products using biotechnology.

These two organizations have established a joint Task force on Orphan Medicinal Products, comprising interested member companies who have either developed or intend to develop orphan medicinal products under Regulation 141/2000. Together, members of the Joint Task force represent a large proportion of orphan drugs currently available on the European market.

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JOINT EBE-EUROPABIO POSITION PAPER
European Commission Public Consultation
Rare Diseases: Europe's Challenges

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

The current EU definition of rare diseases – a prevalence of less than 5 per 10,000 persons – has been in place for a number of years and is fully supported by industry. It recognises the increased complexity of research and development for treatments for rare conditions. Towards the upper end of the scale – as the prevalence begins to approach 5 in 10,000 – it is clear that normal methodologies may be applied to the development of treatments. Therefore, the cut-off point is probably appropriate. Further, it is well-recognised and well-documented (thanks to the EMEA's review of different conditions that have applied for orphan designation). We would suggest that for the review and authorisation process, the current EU definition be maintained.

EBE and EuropaBio would encourage the European Commission to maintain a dialogue with the FDA Office of Orphan Products Development in the US and regulatory bodies in Japan and Australia, with the aim of harmonising the definition globally on this ratio basis. This would be particularly useful now that the Commission and the US have agreed to a common EU/US administrative form for an application for orphan drug status. For this harmonisation process, industry supports the use of the European definition, as it enables the capture of changes in population sizes.

EBE and EuropaBio also support the activities in certain EU Member States in recognition of the specificities of certain rare diseases. Some individual Member States have attempted to address the additional challenges created by increasing rarity by setting specific categories that require special treatment in their healthcare systems. These attempts to find solutions for the issues created by the rarity should be welcomed in the current absence of any EU-level support for the specific needs of very rare conditions. This, however, does not undermine the EU-level definition for review and approval, which should be maintained.

It is crucial to understand and acknowledge that the complexity of all aspects of rare diseases is created by the very fact that they are rare. And that this complexity is a sliding scale, increasing in direct correlation to the increasing rarity. The rarer the condition, and in particular for very rare conditions, the more challenging become all aspects of the treatment, from research, through clinical trials, registration and eventual reimbursement decisions. Commercial uncertainty for the developers of the products also increases with increased rarity.

Question 2: Do you agree that there is a pressing need to improve coding and classification in this area?

Yes.

There is a significant need for better classification and coding of rare diseases and for harmony amongst EU Member States in the identification of people with rare diseases. Such a system should monitor treatment rates, set up epidemiology databases, and would provide a greater understanding of the cause and natural history of these diseases, which would benefit those currently affected and at-risk individuals.

Indeed, many of these diseases are so rare that collaboration must take place at a global level. We therefore agree with the suggestion that the existing WHO International Classification of Diseases should be revised to ensure that rare diseases are adequately coded and traceable in health information systems. At present the WHO-ICD system is not capable of capturing the variety of sub-classifications of rare diseases.

EBE and EuropaBio would also welcome the availability and standardisation of assays / tests with appropriate quality standards.

Question 3: Can a European inventory of rare diseases help your national/regional system to better deal with RD?

Yes.

EBE and EuropaBio believe that a European inventory of rare diseases would help improve both the understanding of the area and the diagnosis of affected individuals, as it would harmonise the data gathering that currently takes place. It would also be of great value to the industry in helping to identify rare diseases, by facilitating understanding of what the situation is with regard to existing treatments and providing accurate epidemiological data.

The Orphanet database, as a searchable database of clinical symptoms, provides information in this regard and is a valuable resource. However, EBE and EuropaBio believe that Orphanet's role should be maintained and strengthened through increased European-level funding, with multi-stakeholder oversight.

A listing in such an inventory – being well-qualified and well-researched – should provide sufficient basis for meeting the prevalence criteria for subsequent orphan medicinal product designation. However, a useful inventory also implies the use of standards for codes and classification (as mentioned in our answer to Question 2).

Further, if a globally-harmonised definition of a rare disease is agreed, EBE and EuropaBio would welcome further harmonisation of this European inventory with other regional inventories, for example, that in the USA.

It is also possible that a European Research Foundation for Rare Diseases would be able to host the European inventory of rare diseases, building on the work and expertise of Orphanet. More details of the proposed Foundation have been set out in our response to question 10 of this consultation.

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

Industry is in complete agreement that there is a need to improve the prevention, diagnosis and care of those who suffer from rare diseases. This position paper has already commented on the value of Orphanet and on the need for continued financial support for the network, so that it can be strengthened and maintained under the auspices of a European Research Foundation for Rare Diseases.

While patient mobility may be necessary to allow a patient to consult an expert in the relevant disease and to gain support from other affected individuals, it should remain in the hands of individual Member States to provide appropriate mechanisms for the optimal treatment and care of people with rare diseases.

EBE and EuropaBio would encourage the exchange of information and of best practice across the EU and it is this that should drive patient mobility. Therefore, we believe that the focus should be on knowledge transfer.

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

Yes.

Modern technology and technological developments should be used to support activities in rare diseases. For example, the use of electronic patient records could be key to patient mobility and allow patients to receive the majority of their treatment close to their home. However, the issue of patient confidentiality must remain high on the agenda, as any data exchanged in this field relates to what is often a vulnerable group of people and to sensitive personal information.

EBE and EuropaBio believe that there may be considerable scope for public/private partnerships in the development of electronic solutions to some of the issues faced and would welcome dialogue with the relevant authorities in this area. The industry would also be open to discussions with the European Commission as to how on-line and electronic tools could facilitate access to information for patients with rare diseases, who may find it difficult to find out the specialised information they need, due to the very fact that their disease is rare.

The European Commission has announced that during 2008 it intends to draft new European legislation proposing to allow a wider spectrum of product information to be provided to patients in the EU. This development is crucial to rare disease patients who, due to the rarity of their condition, are often faced with a long and drawn-out process

before they are correctly diagnosed. Patients may have to wait years – sometimes more than 15 years – for a diagnosis. They may be obliged to travel abroad, again, an option only open to those with the socio-economic possibilities to do so, and may undergo unnecessary or inappropriate treatments, including unnecessary surgery.

Given the nature of rare diseases, patients are often more aware of their condition and the possibility of it being a certain rare condition than their general physician. Currently, companies are excluded from participating in the provision of information to patients on products. Under the new legislative proposals, companies may be invited to play a more active role. If in no other area, this will be a very important development in the field of rare diseases. Given the intense research carried out by companies in the journey to developing a treatment for a rare disease, they often have a wealth of very specific information that could be of use to patients suffering from this condition. They are, therefore, well-placed to participate in the provision of information

It is not clear at this stage what form and format it is proposed that this new information to patients will take, how the information will be verified / monitored, nor what channels will be used to communicate the information. These all remain to be agreed during the course of the development of the legislation. One thing is very clear and that is that this will not include direct-to-consumer advertising. However, for diseases with only very few patients, the internet seems to provide particular advantages as a way of providing and exchanging information. Two factors need to be considered in using the internet as a means of communication – inequalities of access to IT systems, and the need to provide information in all the required languages for EU patients.

Nevertheless, more widespread use of the legally approved documents such as the Patient Information Leaflet (PIL), the Summary of Product Characteristics (SmPC) and the label could form a basis for a non-bureaucratic and cost-effective system.

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

The same high level of quality standards should be applied to rare diseases as applies in any other medical setting, taking into account the rarity of the diseases. The availability and accessibility of accurate diagnostic tests is hampered, as so many other aspects are, by the rarity of the conditions in question. There are simply not enough tests ordered to warrant huge investment in development, validation and certification. Certification and proficiency testing organisations are likely to focus on other areas where the testing is ordered more frequently. This leads to gaps in proficiency testing and means that, however well-meaning, testing is often not accurate.

This is also linked to our answer on Question 9 – if incentives are provided to encourage the development of diagnostic methods, it will be important to make sure that they are used and used correctly, otherwise the incentives will not work in practice.

As long as the field of testing remains a lower priority, the ability of organisations and governments to invest in the field will also remain low. If appropriate screening and

testing were to be more widely explored, investments could be justified in developing and certifying quality tests for rare diseases. European Centres of Reference could play a key role in providing these testing facilities as part of the information-gathering and expertise-building. Given the large number of tests and the need to design and validate a specific set of diagnostic assays for each, no single country can be self-sufficient in the provision of testing. This results in exchange of patient material and testing across national borders. Trans-border flow is clearly a mechanism that will fill a significant gap in the availability of tests for RD. There is a need to enable and facilitate this exchange through clearly stated, transparent, EU agreed quality standards and quality procedures. Regional reference laboratories, properly funded, would be best for channelling samples for the diagnosis of rare diseases. This would also fit well with the “Centres of Reference” concept.

There is an EU-wide quality framework for clinical pathology testing for rare diseases and cooperation between Centres of Excellence, but this is currently on a voluntary basis.

Currently, the EU-funded Network of Excellence EuroGentest (<http://www.eurogentest.org/>) has been set up. EuroGentest is looking at all aspects of genetic testing - Quality Management, Information Databases, Public Health, New Technologies and Education. EuroGentest has been instrumental in making quality assurance procedures and standards known and implemented in the European genetic services and has been providing them support to achieve international quality standards. A common EU platform to harmonize proficiency testing in molecular, cytogenetic and biochemical testing is being created and should be maintained and extended in the future by support from the Commission. Since 70-80 % of rare diseases are genetic or related to the genetic code (cancer), a large part of the work of EuroGentest touches upon rare diseases and genetic testing for them. There is a need for bridging regulatory differences among countries in confidentiality practices, reimbursement, sample transport and storage and accreditation of laboratories. This requires support at the appropriate level (depending on the number of tests per year) to reference laboratories.

Question 7: Do you see a major need in having an EU level assessment of potential population screening for RD?

The consultation document acknowledges the role that population screening can play in helping prevent disabilities in certain conditions. This is not an argument for universal pan-European screening programmes, as these need to be carried out on a national basis, according to the cultural and social values of the country in question. Diagnosis does not automatically lead to automatic treatment. On the contrary, this should be evaluated in each single situation.

Nevertheless, it should be acknowledged that early intervention is the key to successful treatment, especially in the case of progressive conditions. And, the earlier the diagnosis, the earlier treatment can start. Currently, rare disease patients often have to undergo a long diagnostic journey before arriving at a correct diagnosis. This results in high costs for visits, hospitalisation, wrong therapies, including, in some cases, unnecessary surgery.

Depending on the disease, diagnosis by means of a test before clinical signs appear can mean that treatment is initiated before the disease has a chance to cause its clinical burden. This can result in better health outcomes, reduced disability and an increased chance of a healthy life.

New Born Screening programmes, currently in place in some EU Member States, have proven a useful tool to identify genetic diseases. Experience from countries where New Born Screening and other screening programmes are in place should be gathered and best practices – and any benefits accrued from the programmes – should be shared. The European Commission should convene a workshop on the subject to encourage this information exchange.

The European Commission's STRATA recommendations identified genetic testing as a potential tool for the diagnosis and treatment of diseases. It also laid down a series of conditions that should be met for the screening, including genetic counselling. The WHO's criteria laid down in 1965 might not be wholly appropriate in the early 21st Century, given that they were written in a time when the possibility of diagnosis and treatment was a far-off dream. It will be important to ensure that we as a society move forward in light of technological advances, while still holding the key principles intact.

Another question often raised is the value of knowing that one has a condition, if there is no treatment available. Despite the fact that, at the time of diagnosis, there might be no available treatment, giving people information can be useful to help them understand their condition, learn what to expect and manage their condition better for the best outcomes. For example, in France, Cystic Fibrosis is tested for. This allows the best treatment options to be established as soon as possible, to avoid further clinical burden as far as possible.

Additionally, if a moment would come that research into the condition would begin, it could be important for sufferers to be aware of their condition so that they can choose to participate should they so wish. Moreover, an effort should be made to guarantee that all rare diseases for which treatment is available have their specific genetic diagnosis inserted into New Born Screening programmes.

A final benefit to be considered could be the development of increased understanding of disease and disease progression. One of the key issues facing the rare disease community in particular is that there is simply not enough understanding the aetiology of their conditions. A continued evaluation would help researchers to build a fuller understanding of what they are, what causes them and – maybe more importantly – how they are caused. This, in turn, could support therapy development by helping us understand how therapies can or could intervene in the process.

One important aspect is that the Paediatrics Regulation requires including paediatric subsets. A waiver cannot be requested based on the fact that the disease is not diagnosed

in the paediatric population. Therefore, testing should be used to establish such clinical subsets that might require further research.

Therefore, although possibly ambitious, the combination of New Born Screening together with registries and – potentially – a national fund for rare diseases managed at a central level, could significantly increase the information (epidemiological, clinical evolution, etc.) available about these pathologies.

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

Accessibility to Orphan treatments is one of the most pressing issues in the rare disease field. Before the creation of the Orphan Regulation, the main problem facing rare disease patients was the lack of treatments. Today, even where treatments are available and authorised, patients are still being denied those treatments. Successful access often comes following political pressure, legal challenges and persistent action. This tends to favour the well-connected, socially and educationally advantaged members of our EU society. This is clearly contrary to the spirit of the Orphan Regulation, which is based on the principles of societal equity and solidarity.

As healthcare systems remain under the responsibility of Member States, it is, ultimately, the Member States that will make the decisions determining the availability – or not – of a treatment. Nevertheless, we do believe that there are several activities at an EU level that can support availability of orphan treatments, provided information and understanding of the orphan system can be increased.

Possible solutions for addressing the access problem will need to be addressed by cooperation between all stakeholders. However, our suggestions are as follows:

1. Raise awareness of the functioning of the Orphan system, particularly amongst payers and decision-makers in the Member States. This should include awareness of the unique provision, that Orphans are required to prove that there is either no existing satisfactory treatment or that the new one is clinically superior.
2. Member States should acknowledge the expert decisions taken by their representatives on the COMP and the CHMP.
3. An acknowledgement should be made that for conditions below a certain prevalence, normal Health Technology Assessment methodologies will always be inappropriate, simply due to the rarity of the disease. The COMP – as the expert body made up of Member States' representatives reviewing the prevalence data – should recommend if this is the case or not. This also reflects the heterogeneity of the orphan field.
4. Orphan medicinal products falling into this category should be approved for reimbursement in the Member States following the granting of a Marketing Authorisation, on the basis that they are too rare to be able to follow HTA rules and on the fulfilment of the EU legal requirement to have proven either that no other treatment exists or that they offer clinical superiority.

5. If this is the case, sponsors should be required to gather experience from the field in order to develop the data sets – including clinical effectiveness and outcomes – which should be submitted to the reimbursement authorities at pre-determined points. These should be realistic according to the rarity of the disease. However, complexity increases in correlation to rarity. The system should reflect this sliding scale of complexity, allowing for thorough examination on a case-by-case basis. For rarer diseases, therefore, the review points of the data gathered should be set further out to allow for the development of meaningful data.

We also welcome the proposal for the Commission to present a regular report to the Council and the Parliament identifying bottlenecks (delays, marketing, access, reimbursement, prices etc.) and proposing any necessary legislative modifications in order to guarantee equal access to OMPs throughout the EU. This reporting should be based on a broad consultation with physicians, industry, patients and other relevant stakeholders. To ensure a balanced view, the Working Group of Interested Parties at the COMP should support this process.

We also agree that a better system is needed to provide necessary medicines to patients in need before approval and/or reimbursement is granted. Compassionate use is an important tool to provide patients access to treatment in the case of severe and urgent medical need. And, while compassionate use programmes should not be used to replace participation in a trial, the urgent and life-threatening nature of many rare conditions should be recognised. Administrative procedures can delay access to treatments. Where a positive risk/benefit profile can be assumed – for example where the patient’s life is in immediate danger – it may be an appropriate decision to allow early access to the treatment under development to the patients with rare diseases.

Compassionate use programmes should not be used to replace participation of patients in a clinical trial but to enable fast access to treatments for patients with rare diseases as long as the product is not approved. Stakeholders are called to minimize the administrative procedures and ensure expeditious reimbursement of the orphan medicinal products once marketing authorization has been granted.

An improved European system should promote transparent and equal access to compassionate use, based on patient needs rather than socio-economic advantages or political or social connections.

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

Rare disease patients deserve the same access to safe, efficacious and high-quality treatments as patients with more common conditions. We believe that the importance of accurate diagnosis for patients with rare diseases should be recognised. Therefore, we would support the creation of incentives or extension of existing incentives to the developers of medical devices and diagnostics.

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

Social and educational services for people affected by rare diseases are important and EBE and EuropaBio agree that resources should be provided to support such activities. Given the specificities of culture, language and lifestyle, we would propose that each individual Member State determines the services it will provide for rare diseases patients and care-givers.

However, at an EU level, we believe that the private sector may have significant contributions to make in delivering educational services to the rare disease community. A company developing a treatment will have particular knowledge of a given disease and the Commission should consider how this knowledge and expertise can be shared effectively with healthcare professionals and those affected by the disease.

As mentioned previously, EBE and EuropaBio support the Commission's proposal to link international (European) databases to national and / or regional databases. We would also recommend linking these to databases outside of the European Union, which would increase population sizes and facilitate the study of some of the rarest of rare diseases.

Question 11: What model of governance and of funding scheme would be appropriate for registries, databases and bio-banks?

Registries can play an important role in capturing the data suggested by our proposals under Question 8. Several registries exist already. In order to make the most of the investment in the registries, the data should be used in the most effective way to provide stakeholders and decision-makers with information that could be lacking due to the rarity of the conditions in question.

One key aspect is the need for collaboration. At a minimum, registries should be established at a European, rather than national level. This will allow for the development of a reliable sized dataset for analysis. The Registries will, anyway, allow data analysis specific to countries as it could be an interesting contribution for generating robust epidemiological data for the decision-makers.

EU support, funding and regulation should be considered for establishing the basic underlying elements to set up these data banks at Reference Centres for specific rare diseases.

Public/private partnerships would then be the appropriate way for them to be further developed and governed. However, informed consent and data privacy must be given due regard in their development. Equally, EBE and EuropaBio would encourage a discussion as to who would have access to such information. A number of ethics guidelines for the use of human tissue have been developed and these should also be adhered to.

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?

Industry and charities have a vital role to play in the development of an EU action on rare diseases and must be recognised as partners of equal value. The common interest of all stakeholders in seeking to ensure that patients have access to early diagnosis and effective treatment, is best served by working in partnership with public bodies.

We believe that industry has a crucial role to play in the development of information to rare disease patients on their condition and treatment options. This should not be direct-to-consumer advertising, but should tap into the information developed by companies and their researchers during the development of a treatment for a rare condition. We look forward to the European Commission's proposals for legislation in this field during the course of 2008.

One of the most effective tools in the EU has been the establishment of multi-stakeholder think tanks or steering committees to advise governments and decision-makers on the best way forward in the field of rare diseases in their country. Observations have indicated that rare disease patients are better served in countries where such multi-stakeholder groups exist.

We are concerned that industry should be – and is currently not – viewed as a partner of equal value. One concrete example is the exclusion of industry from the drafting group that prepared this consultation. Although we welcome the opportunity to participate in the public part of the consultation exercise to which we are now contributing, we would seek that industry be viewed and treated as a partner of equal value in the process.

The COMP Working Group with Interested Parties (COMP WGIP) is a multi-stakeholder consultation group that could fulfil this process and should be used to fuller extent in situations such as this.

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?

Yes, at a national level.

We strongly support the idea of having action plans and believe that these can best be developed and implemented at a Member State level. Regionalisation of healthcare is a challenge for rare disease patients and, wherever possible, we believe the challenges of rare diseases can best be handled at a national level.

We have seen value in the use of national action plans in other therapeutic areas and would like to see national action plans or other strategic approaches developed in the field of rare diseases – with the proper consultation of all stakeholders.

EBE and EuropaBio would welcome the coordination of these action plans by the European Commission. We support the concept of a regular review and reporting by the Member States on the development and implementation of the action plans as this will encourage the sharing of best practice and creative ideas. In this way, they would be useful for benchmarking and sharing best practice at Member state level.

We would argue that the national plans should first and foremost see the creation of a central fund for giving patients access to the treatments already available. They should also foresee the creation of multi-stakeholder steering groups to guide decision-makers in the effective treatment for rare disease patients and their families.

The outcomes of such National Plans should form part of the Commission's regular reporting on the progress being made in the field, particularly focussing on bottlenecks to access.

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

No.

EuropaBio and EBE do not see the need for a new European Agency: we believe that the existing structures have the knowledge, experience and expertise to progress any actions agreed as a result of this consultation. The establishment of a European Agency would require time-consuming procedures and would divert resources away from the pressing needs of the rare disease community.

However, we do support the establishment of a European Research Foundation for Rare Diseases. The aims of such a Foundation should include:

- Pooling of the wide-spread central funding efforts of the European Union for rare diseases to allow more rational and aligned budget decisions;
- Initiation and support of cutting-edge biological research to discover the causes of diseases, find and improve methods of prevention and develop more effective diagnoses and new treatments;
- Initiation and support for the set-up of pan-European patient registries, natural disease databases and networks of excellence;
- Support and advice for Member States in the set-up and implementation of national action plans on rare diseases;
- Regular organisation of international conferences and congresses on rare diseases; and
- Consultative status to the European institutions, including high-level scientific and policy support in international harmonisation efforts.

The European Research Foundation for Rare Diseases would have a range of diverse teams, with scientists investigating all aspects of the disease process, from understanding genetic changes to the range of phenotypes which might arise from these changes and the different treatment strategies that are needed.

EBE and EuropaBio believe that such a Research Foundation is best achieved by a governance structure that includes all interested stakeholders, including people affected by rare diseases.

Prior to launching a feasibility study in 2009, we would like to suggest to the Commission to use the opportunity of a conference on rare diseases in September 2008 in Paris to dedicate a multi-stakeholder workshop on this topic. If deemed useful, EBE and EuropaBio would be happy to support such a workshop by bringing our expertise to an organising committee.