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- German Alliance for Rare Diseases –

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Comments on the Public consultation

"Rare Diseases: Europe´s Challenges"

EUROPEAN COMMISSION
HEALTH & CONSUMER PROTECTION DIRECTORATE-GENERAL
Directorate C - Public Health and Risk Assessment

0. **General remarks**
   ACHSE is the umbrella organisation of patient organisations for rare diseases in Germany. We comprise more than 70 patient organisations representing about 1,000 rare diseases in our country. ACHSE is recognised as a "National Alliance" by the European Organisation for Rare Diseases (EURORDIS).
   We explicitly welcome this initiative of DG SANCO and the opportunity to comment on that public consultation. This further enhances EU's policy for rare diseases which was promoted significantly by patient organisations in the past.

   To facilitate the identification of research and public-health projects funded by the EU, we suggest to ask applicants generally to state whether and how the project is of relevance for rare diseases. The answers may help applicants to think about rare diseases within their medical speciality and potential links to the project they apply for.

1. **Q1: Is the current EU definition of a rare disease satisfactory?**
   Yes, we strongly support the current definition as laid down in directive 141/2000, i.e. less than 5 per 10.000 persons in the EU. As already mentioned in the text, the majority of MS is applying that definition that has proven its feasibility. Further discriminating between different "rare" and "very rare" diseases would jeopardize the current efforts to improve both diagnosis and therapeutic care for

the entire rare-disease population.

2. **Q2: Do you agree that there is a pressing need to improve coding and classification in this area?**
   Yes, this may be principally useful, however, any resources devoted to this effort should not be taken from programmes directed to improve diagnosis and care directly. While a better coding may be helpful for some scientific purposes, we doubt the immediate benefit to patients of an EU inventory of RD. We suggest involving umbrella patient organisations to learn about actual problems with coding and classification that have a direct impact on health-care services to patients.

3. **Q3: Can a European inventory of rare diseases help your national/regional system to better deal with RD?**
   Undoubtedly, an inventory as described in the text would be a very useful tool, especially for researchers. However, we doubt that it will be realistic to get reliable information about prevalence and aetiology for all rare diseases in the EU. We prefer a bottom-up approach over the suggested top-down concept. Specifically, we recommend to set up an anonymised inventory of undiagnosed cases. In that kind of inventory, symptoms and syndromes should be compiled for which no diagnoses could be found. This may help not only to improve knowledge about rare diseases in general but also to find diagnoses for yet undiagnosed patients.

4. **Q4: Should the European Reference Networks privilege the transfer of knowledge? The mobility of patients? Both? How?**
   The rarer a disease and the smaller the MS is, the more it becomes unlikely for patients to find appropriate in- and out-patient care in his/her home country. However, cross-border care adds complexity such as language, cultural and reimbursement issues and should therefore be given lower priority than the transfer of knowledge between expert centres within a disease-specific network. We welcome any efforts to set up EU-wide networks of reference/expertise if these networks are devoted primarily to make the optimal diagnostic pathways and/or therapeutic options available to patients both directly and indirectly via associated regional centres. Over and above this objective, the networks should facilitate both pre-clinical and clinical research. It will be important to ensure a patient-oriented approach including psycho-social aspects and co-operation with qualified patient organisations.

5. **Q5: Should on-line and electronic tools be implemented in this area?**
   Finding reliable information about rare diseases that is both up to date and understood by a lay audience remains an important challenge to patients. ACHSE has therefore developed the concept of patient-oriented disease descriptions that have to meet certain quality criteria. Applied to an internet-based search portal this concept facilitates the identification of quality-assured information about rare diseases. We offer support to extend this concept outside Germany. Certainly, we support in addition online- and/or electronic-tools that facilitate finding a diagnosis for yet undiagnosed patients. In addition, we suggest to establish and operate an on-line scientific journal for case-reports on rare diseases or syndroms, both diagnosed and undiagnosed. Unfortunately, such case-reports are

not well accepted in scientific journals although they may be extremely helpful to foster progress in research.
ACHSE also supports the harmonisation of electronic patient records that can be a useful tool to exchange information on rare disease patients within the EU.

6. **Q6: What can be done to further improve access to quality testing for RD?**
   We support any efforts to improve the level of evidence for diagnostic procedures and the exchange of information about these procedures. In particular, EU recommendations for minimum standards may be helpful. However, the principle of subsidiarity should be observed.

7. **Q7: Do you see a major need in having an EU level assessment of potential population screening for RD?**
   It must be clearly defined what the EU means by "population screening". ACHSE supports post-natal screening services for those diseases that can be reliably screened, i.e. with both sensitivity and specifity appropriately optimised, and for which an impact of early post-natal diagnosis on the application of effective curative or symptomatic treatment has been shown. It is important that the actual accessibility of treatment options rather than their theoretical availability is taken into account in this context. We oppose post-natal screening for diseases for which treatment is either not accessible to the majority of patients or not available at all. We reject population screening for gene carriers who are not phenotypically affected and pre-natal screening unless in-utero treatment is available. As ACHSE is devoted to fight the diseases rather than those affected by them, we cannot see the advantage of identifying diseased embryos or foetuses if the only option left would be terminating the pregnancy. Under no circumstances, individuals should be directly or indirectly obliged to disclose results of screening test to any third party including but not limited to insurance companies and/or public insurance schemes.
   Having said this, recommending rather than legally setting the minimum criteria for population-wide screening for defined RD may be useful as it potentially helps to improve access to screening in the MS.

8. **Q8: Do you envisage the solution to the orphan drugs accessibility problem on a national scale or on an EU scale?**
   From a standpoint of patient solidarity across Europe, we certainly welcome all efforts encouraging true and equal accessibility to all treatment options including drugs all over Europe. However, social security systems are for good reason within the remit of the MS. We do not consider it useful to extend EU's responsibilities to this area and favour therefore to solve accessibility issues on a national scale.

9. **Q9: Should the EU have an orphan regulation on medical devices and diagnostics?**
   Although some important differences between medical devices and orphan drugs exist, i.e. medical devices are less disease specific than orphan drugs and therefore may be usable in a wider range of patients, we do realize that an approach comparable to orphan drugs may be useful to patients. For example, there is no doubt that certain in-vitro diagnostics are specialized for the use in rare-disease patients and we therefore welcome any plans to extend the orphan-

drug regulation concept to diagnostics.

10. **Q10: What kind of specialised social and educational services for RD patients and their families should be recommended at EU level an at national level?**
    Health Technology Assessments of Orphan Drugs on an EU level may help to foster implementation of these drugs into national reimbursement schemes. However, we realize that only the benefit/risk ratio rather than the economic component can be reliably assessed on an EU level given the diversity of social security systems. We welcome efforts to harmonize compassionate use programmes, e.g. by means of guidance provided by EMEA and/or common policies agreed between MS facilitating the implementation of such programmes on a national base.

    Social support such as respite care services, information services and help lines as well as therapeutic programmes may be applied across member states to the benefit of patients if such services cannot be provided in the respective MS itself. However, language and cultural barriers have to be taken into account. We therefore are more in favour of empowering member states and non-governmental organisations in providing such services directly to patients in the MS locally.

11. **Q11: What model of governance and of funding scheme would be appropriate for registries, databases and biobanks?**
    Rare diseases are for obvious reasons perfect models where a European setting is superior over a national approach when establishing registries, databases and biobanks. The legal framework (data-protection, good practices) should be already implemented on the European level and can therefore be used readily. Ownership of both material and data is critical to ensure access to these resources for all scientific groups that are devoted to patient-oriented research. As funding is often the key to ensure ownership and to enforce true and fair access, we strongly favour either exclusive public (i.e. national and EU-funding combined) funding or partnerships between non-commercial and public institutions. Potential partners comprise charities, patient organisations and public research funding agencies including but not limited to EU-commission`s institutions. A steering committee should oversee and govern the policies of the specific projects. In this committee representatives of all funding parties, of scientists and of patient organisations should be represented. We are concerned about commercial sponsors claiming ownership over the material and information and limiting access to it. We dislike, however, also any efforts of single scientific groups to set up biobanks, registries and databases in an non-inclusive manner, i.e. excluding other scientific groups from accessing the resources from which everybody could potentially benefit.

12. **Q12: How do you see the role of partners (industry and charities) in an EU action on rare diseases. What model would be the most appropriate?**
    The co-operation of the different stake-holders in rare-disease research is absolutely critical to bring true progress to patients. Neither can research efforts be left alone to the pharmaceutical industry that is likely to apply a cherry-picking policy that favours medical fields with a high likelihood of a good return on

investment nor will the public and civil sector be sufficient to bring together knowledge and financial resources needed to develop new therapies. It will therefore be key to bring together industry, patient-organisations, philanthropic organisations (e.g. private foundations), academic centres and public funding agencies to ensure that programmes are set up that are directed to the most burning unmet medical needs of patients living with a rare disease. Good ways to ensure such co-operations are matching grants offered by public agencies and charities jointly. Such programmes will provide incentives to commercial partners that are willing to take a share of the development risk. Likewise, tenders for bids of research and development work can be offered by an EU agency on rare diseases. Partners can bid for these tenders by offering skills and/or financial resources becoming thereby governing partners of the specific development projects.

The co-operation of industry, patient-organisations, other charities and public institutions must be based on the principles of transparency for all partners involved and the independence of patient organisation. We clearly oppose any attempts to abuse patient-organisations in a hidden agenda for third-party interests.

13. **Q13: Do you agree with the idea of having action plans? If yes, should it be at national or regional level in your country?**
We strongly favour an action plan for rare diseases in Germany and encourage other MS to follow the example of France and a few other European countries. A national plan for rare diseases that helps to integrate all efforts directed to improve diagnosis, care and research related to these diseases. Such a plan, however, will be only a major step forward if it is implemented effectively. As Germany is a federal state, a national plan must be set up by both the Federation and the states (Länder) to ensure that really all relevant areas are covered.

14. **Do you consider it necessary to establish a new European Agency on RD and to launch a feasibility study in 2009?**
As already mentioned earlier, actions integrating the research for and the evaluation of new diagnostic and therapeutic options for rare disease as well as co-ordination of national efforts on the basis of the subsidiarity principle should be concentrated at a single institution on EU-level. An RD-agency can also provide advice to academic groups, industry and patient organisations on organisational and scientific problems related to rare diseases in common. ACHSE is therefore in favour of establishing a European Agency on Rare Disease. The office for rare diseases of the National Institutes of Health (NIH) may act as an example, how such a structure can be established in a federative context. We appreciate concerns about additional bureaucracy and therefore suggest to focus on the co-ordinating function of the RD-agency. Patient organisations must be represented in the steering committee and applicable working parties, groups or committees of the RD-agency.

Berlin, 13th February 2008