Deutsche Sarkoidose-Vereinigung e.V.

- German Sarcoidosis Association –

Comments on the Public consultation „Rare Diseases: Europe's Challenges"

DG SANCO – Directorate C

0. General remarks

The ‚Deutsche Sarkoidose-Vereinigung e.V.' (DSV) is the association of people suffering from sarcoidosis and other granulomatous disorders, fibrosis and other rare diffuse lung diseases. All of these diseases are rare disorders. The association was founded in 1987 and has actually 4.200 members in Germany. It cooperates with partner associations in other European countries and their European ambrella organisation, EPOS (European Association of Patients Organizations of Sarcoidosis and other Granulomatous Disorders). DSV is member of ACHSE and EURORDIS.

As a patient association we explicitly welcome this initiative of DG SANCO and the opportunity to comment on that public consultation. The patients in Europe need further enhancements concerning rare diseases like Sarcoidosis.

Sarcoidosis itself covers several ICD-10 codes and consists of several sub types classifying the course of the disease, such as acute sarcoidosis (Loefgren's Syndrome), chronic sarcoidosis, recurrent Loefgren's Syndrome, and consists of different sub types concerning the pattern of the organ involvement, like e.g. Loefgren's Syndrome, Morbus Jüngling, Sjoegren's Syndrome, Heerfordt's Syndrome, Lupus Pernio, juvenile sarcoidosis (sarcoidosis in childhood) or berylliosis.

The morphological characteristic is the chronic inflammation with the development of granulomas which can develop in nearly all organs causing loss of organ functionality and irreversible damage if the disease develops progressively. Therefore sarcoidosis can mimic numerous other diseases and need proper differential diagnosis. Because the aetiology is unknown yet, a causual therapy is missing.

Thus basic research and clinical research is needed. This should be coordinated Europe wide and be supported by MS as well as EU.

In order to strengthen synergy effects we would like to ask for the introduction of a feature into the application process of medical research and public-health projects funded by the EU that applicants generally state whether and how the project is of relevance for rare diseases. The answer may help applicants to think about rare diseases within their medical efforts and identify links to opportunities for rare

diseases of the project they apply for.

We were missing a question concerning programmes to support patient organisations in Europe, e.g. to support the co-operation of patient associations in MS within a group of rare disorders.

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. 5 per 10.000 persons in the EU. The majority of MS, and also countries abroad, is applying that definition that has proven its feasibility. Discriminating between different kinds of “rare” and “very rare” diseases would jeopardize the current efforts to improve both diagnosis and therapeutic care for the entire rare-disease population.

Q2: Do you agree that there is a pressing need to improve coding and classification in this area?
Yes, because the systematic of diseases by improving classification may be helpful in basic research, epidemiological research and research for the cause of sarcoidosis. But ressources for coding improvements should not be taken from programmes directed to improve diagnosis and care directly. We would like to give input as patient association also concerning problems with coding and classification.

Q3: Can a European inventory of rare diseases help your national/regional system to better deal with RD?
Yes, an inventory as described in the text would be a useful tool for researchers. It would be helpful also for epidemiological progress. Most efficient to work we expect a bottom-up approach.

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 the home country.

This is especially true also for different organ involvements of sarcoidosis, like e.g. neurosarcoidosis or lupus pernio (sarcoidosis of the nose) or sarcoidosis of the upper airways or heart sarcoidosis etc. as well as special forms like berylliosis, a form where the cause (beryllium exposure) could be detected. So for special patient groups also individual travel of patients should be possible, but in general transfer of knowledge would be preferred.

Because cross-border care increases in general the 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 e.g. via associated regional centres. Moreover 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.

Q5: Should on-line and electronic tools be implemented in this area?
To find reliable information about sarcoidosis and rare diseases which are reliable and easily understood is an open problem. Public access to scientific resources like Cochrane reviews etc. would improve the access to reliable information, but additional projects are needed to provide such reliable information to patients in Europe they can understand.

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, as usual based on the principle of subsidiarity.

Q7: Do you see a major need in having an EU level assessment of potential population screening for RD?
The answer depends on how EU understands “population screening” in detail. In general a post-natal screening service should be implemented for diseases for which an effective curative or symptomatic treatment exists and is accessible.

We do not support general population screening for gene carriers who are not phenotypically affected and pre-natal screening unless in-utero treatment is available.

Q8: Do you envisage the solution to the orphan drugs accessibility problem on a national scale or on an EU scale?
Because sarcoidosis is a world-wide disease we certainly welcome all efforts encouraging true and equal accessibility to all treatment options including drugs all over Europe. Despite the national differences of the health care systems in the MS the market for Orphan Medical Products may be handled better on European level,

because of expertise on this level and in order to overcome access problems by national reimbursement regulations.

Q9: Should the EU have an orphan regulation on medical devices and diagnostics?
Yes, we welcome any plans to extend the orphan-drug regulation concept to diagnostics or medical devices which may be useful to patients with rare disorders, also when patients not only of one disease benefit, but patients with several diseases.

Also 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.

Q10: What kind of specialised social and educational services for RD patients and their families should be recommended at EU level and at national level?
Social support such as respite care services, information or educational 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.

Q11: What model of governance and of funding scheme would be appropriate for registries, databases and biobanks?
For rare diseases such as sarcoidosis 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.

Because funding is often the key to ensure ownership and to enforce fair access, we favour either exclusive public (i.e. national and EU-funding combined) funding or partnerships between non-commercial and public 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.

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?
As a lot of examples have shown, the co-operation of the different stake-holders in rare-disease research is the key to achieve real progress to patients. Industry focus alone means that pharmaceutical industry is likely to concentrate solely to diseases with quick and optimal return on investment. Purely public driven can hardly bring together knowledge and financial resources needed to develop new therapies.

Thus a coordinated approach bringing together industry, private foundations, academic centres, public funding agencies and patient-organisations should face the unmet medical needs of patients living with a rare disease.

As a rule, the co-operation of industry, patient-organisations, other charities and public institutions must be based on the principles of transparency and independence of all partners involved.

Q13: Do you agree with the idea of having action plans? If yes, should it be at national or regional level in your country?
Yes, 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. This should be within an European framework.

Q14: Do you consider it necessary to establish a new European Agency on RD and to launch a feasibility study in 2009?
Yes, a European coordination of the efforts on RD is essential as a continuing task and not from one project period to another.

Such an RD agency should provide advice to academic groups, industry and patient organisations on organisational and scientific problems related to rare diseases in common. Patient organisations must be represented in the steering committee and should be involved in working committees of the RD agency.

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