Osteogenesis Imperfecta Federation Europe

(OIFE)

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

DG SANCO – Directorate C

Bamberg, February 13th, 2008

General remarks

O Osteogenesis imperfecta (called „OI" or „brittle bones") is a rare, hereditary disease of the connective tissue. One of its main symptoms is a high fragility of the bones. Other possible symptoms that might occur are for instance: growth deficiency, hearing loss, deformities of the long bones and the skeleton, loose joints or fragile teeth (dentinogenesis imperfecta). OI has an incidence of around 1 : 15.000, is caused by a lack or a structural defect of collagene and shows a large variety of symptoms.

The **OIFE** is the European Federation of national OI associations, founded in 1993. Actually it has 15 European full members and six associate members from Australia, Ecuador, Georgia, Mexico, Peru und Northern America. The OIFE is registered in the Netherlands and actually has its European office in Bamberg/Germany since 2001.

Q1: Is the current EU definition of a rare disease satisfactory?

Yes, we have worked with this definition in different countries and organisations of different structures and found it completely sufficient

Q2: Do you agree that there is a pressing need to improve coding and classification in this area?

Yes, this may be helpful, but for us other for us other aims (with a more practical and direct impact on daily life of OI-people and their families) have a much higher priority. An early diagnosis and adequate care for OI-people in all European countries is much more important.

Q3: Can a European inventory of rare diseases help your national/regional system to better deal with RD?

OIFE is a European Federation, but as far as we know from our various members. Such an inventory would certainly be useful for researchers.

To bring this through we would prefer to start with such inventories in every country and with an immediate combination of these data throughout Europe.

Q4: Should the European Reference Networks privilege the transfer of knowledge? The mobility of patients? Both? How?

For people with a rare disease as OI it is often very difficult to find appropriate in- and out-patient care in one's home town or often not even in the home country.

The possibility to be treated in another country should be available, but in reality this is difficult, not only for financial reasons. Therefore the transfer of knowledge between experts from different countries should be given a much higher priority and in our opinion it is more efficient and can bring more benefit than the transfer of individual patients.

We regard it as highly necessary to set up a close cooperation between scientific and medical networks and qualified patient organisations.

Q5: Should on-line and electronic tools be implemented in this area?
Yes, of course, as they are more easily available both for patients as for researchers and professionals!

Q6: What can be done to further improve access to quality testing for RD?
We see the need of free access to diagnostic procedures in all EU-countries and for better qualified medical professionals.
In case of Rare diseases it can not be expected of every clinic and every doctor to possess all information, but in case of doubt the transfer to another clinic must be easy to attain and financially secured.

Q7: Do you see a major need in having an EU level assessment of potential population screening for RD?
That depends on the definition of "population screening". If a screening will have an effect on the availability of treatment and care for disabled people, we welcome this concept. But we oppose 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?
From the view of an international Federation like us, we certainly welcome all efforts encouraging true and equal accessibility to all treatment options including drugs all over Europe. But EU-authorities should not interfere with national health or social security systems apart from supporting countries to learn from each other in order to make sure that a sufficient and similar standard of treatment will soon be available in all EU-countries.

Q9: Should the EU have an orphan regulation on medical devices and diagnostics?
If that will help to improve the accessibility of medical devices and diagnostics in all EU-countries, yes!

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?
Integration of disabled citizens, independent living and full participation are of first importance for all RD patients. But whereas we are generally in favour of an integrated approach of education for disabled people we would welcome special support or care services, information services and help lines as well as therapeutic programmes if for some disabled people the participation in regular schools and education is not easily possible.

Q11: What model of governance and of funding scheme would be appropriate for registries, databases and biobanks?
Especially in rare diseases a European cooperation is necessary when numbers in a national registry would be too small. The legal framework (data-protection, good practices) should come from a European level, funding should be connected to an internationally and interdisciplinary team/committee including representatives from research institutes, patient organisations and non-commercial sponsors.

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?
In our opinion the support of these partners is very welcome and necessary, but the cooperation of these organisations should be organised by a committee with representatives of each partner and the independence of all partners must be secured and guarded. Apart from this the interests, motivations

of and benefit for each partner must be made transparent.

Q13: Do you agree with the idea of having action plans? If yes, should it be at national or regional level in your country?

Of course such an action plan is necessary and - if possible – should be based on a national level. If regional sub-groups or organisations act independently or if a country should have several umbrellas for rare diseases (which is not favourable in our opinion, but does occur!) they should be requested to cooperate closely and to avoid competition or conflicting actions between each other.

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

Yes, we regard this as highly necessary to encourage research for and the evaluation of new diagnostic and therapeutic options for rare diseases.
Such a European Agency on Rare Disease should and could coordinate actions and help to avoid double activities in the same field. Certainly Patient organisations should be represented in the steering committee and applicable working partys, groups or committees of the RD-agency.

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