EC consultation paper: „Rare diseases: Europe's challenges"

- Comments by Retina International –

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1. Patient Empowerment:

The new paradigm of „patient empowerment", mentioned in the EC paper, should have been its "fil rouge", the main theme streamlining all topics related to rare diseases. As patients and relatives affected by rare diseases ("RD patients"), in particular very rare disease patients, are often the only "experts" of their disease, their individual and collective experience must be considered the most valuable input shaping aspects of RD related health care and research. Patient empowerment is the provision of resources which permit equally affected patients, i. e. RD patients with identical medical diagnosis, to pursue their own health related objectives. Patient empowerment therefore, is not only based on external support, but equally on mutual support by peers. The EC should be aware that a rare disease initiative without stringent patient orientation will fail. Patient groups are not, as stated in the communication, "among the most empowered groups" in the health sector. In order to reflect the utmost importance the RD patient empowerment it is suggested that the small "p" is introduced as a new suffix to designate concepts, activities, guidelines, projects etc. determined by or oriented towards RD patients –similar to the small "e" reflecting the electronic paradigm. Analogous to e-business reflecting the pursuasive importance of electronic tools in modern business, "p-health" reflects a patient centred health care system.
The following comments are based on this rather "radical" patient perspective which is dictated by the special chronic needs of (very) rare disease patients.

2. Question 2/ Question 3: Do you agree that there is a pressing need to impose coding and classification in the area of rare diseases? Can a European inventory of rare diseases help?

The German umbrella organization for rare diseases, ACHSE, has developped a list of about 20 p-criteria for describing rare diseases such that patients can best profit from this information.
A patient-oriented coding and classification system ("p-classification") should reflect the function of such a classification for the patients themselves. The name of the disease and the exact diagnosis should not only convey its medical connotation, but should be comprehensible to the patient and his/her relatives. The patient needs to understand the rare disease in order to talk about it to peers, relatives, carers and doctors. Also the designation of a rare disease has a social function in group formation, helping RD patients to find equally affected peers and looking for disease-related information.
2 examples: The rare retinal degenerative disease "Gyrate atrophy" is due to "Hyperornithinemia". A p-description of this disease should contain special information about how to reduce the abnormal ornithin plasma level by diet as well as vitamin B6 treatment. Such a p-disease-description of Gyrate Atrophy would also focus on the negative impact on the quality of life of an amino acid free diet compared to the comfortable intake of pills with vitamin B6.
Such a p-classification also has positive consequences for statistics and data bases established by patient organizations helping them to demonstrate to the public their experience in supporting and managing the respective rare disease patient subgroups. A genetics-based **sub**-classification should be considered, in order to orient patients towards upcoming gene-based therapy trials.

Similarly the name Retinitis Pigmentosa should be used rather than Retinopathia pigmentosa for the blinding retinal degenerative disease, since these designations are generally applied by patients and form part of their associations' logo (e.g. British Retinitis Pigmentosa Society).

3. Question 4: Should the European reference networks privilege the transfer of knowledge? The mobility of patients? Both? How?

Eurordis has recently organised a number of national and international workshops with patient organisations and other stakeholders, defining and analyzing criteria and functions for ERNs which should be taken into account. Quality criteria for medical and all-comprehensive socio-psychological services are paramount and transfer of knowledge between ERNshould be preferred to patient mobility since language obstacles and severe mobility restrictions hamper cross-border movement of RD patients.

p-clinical services should be established in RD clinics in close cooperation with RD patient groups, including specialized access and information services, direct contact with representatives of patient organisations, on-site peer counselling, bedside services. Outcome results of clinics should be made publicly available in order to enhance informed patient choices, and RD expert recruitment and expert training should take experience of patient organizations (PO) into account.

4. Question 5: Should online and electronic tools be implemented in this area?

The experience of RD patients in the management of their disease and about effect of treatments is often documented in paperbound records or PC dossiers. Most RD patients are interested on health records and should be encouraged to develop these further and make use of p-electronic health records; support and training services for RD patients and their PO should be established to provide and profit from access to these new electronic and web-based tools. Databases and studies on best practice models in this area should be made available and adapted to the needs of RD patients, including record transfer between doctors and RD patients.

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

Diagnosis and treatment of RD patients is often delayed, because the doctor of first contact is not sufficiently informed and has no or only scarce experience with the RD. In order to improve the RD patients chances of receiving early and accurate diagnosis and treatment it is recommended that the patient-doctor dialogue is improved by model initiatives on a national and European level:

- Further training of doctors and education of medical students by qualified representatives of PO in RD
- Establishment and support of “escort services” of PO so that family members and peers may accompany RD patients during their visits to doctors
- Dissemination of DVDs for doctors and for use in doctor training courses demonstrating an ideal “trialogue” between RD patient, representative of PO and the doctor. This “trialogue model” may help to broadly introduce collective RD patient experience into the doctor-patient contacts.

6. Question 12: How do you see the role of partners in an EU action on rare diseases? What model would be the most appropriate?

Public-private partnership between EC, industry and charites/PO should be in the center of future RD research funding models. The most important first step in the establishment of such partnerships is to provide a data basis on funding activities of RD patient organizations as well as information system on research projects initiated, funded, coordinated and

disseminated by RD patient groups or umbrella organisations. They could provide the basis for effective public private partnerships based on the authentic research needs of RD patients. POs also provide (co-)funding for professorships, laboratories, post doc fellowships, colloquia, DNA chips, biomarker banks and other projects largely unknown to the EC and the public.
Patients should be represented in RD projects in research advisory boards of the EC and ERF, and projects initiated by patient organisations should be co-funded by 100% by EC/MS. In particular the establishment and administration of data bases, websites and registries where POs have ample experience of their own, should take patient organizations expertise on board.

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

We indeed fully support the idea of such a specialized EU agency for rare diseases, since (quotation from the EC paper): “An EU Agency dedicated to RD can be an excellent instrument to insure the permanence and coherence of relevant strategies at EU level in different areas such as patient registries, bio banks, clinical trials, information on RD, networks of centres of reference, consensus clinical care recommendations and quality assessment”. Also it should be partner of national and European RD patient alliances who should be represented on its main decision making and advisory boards. In order to provide synergy effects with non-RD disease areas, also representatives from other European disease-specific patient umbrella organisations should be taken into account (e. g. EPF).