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Rare Diseases Europe

POSITION PAPER

EUROPEAN REFERENCE NETWORKS FOR RARE DISEASES

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Rare disease patients have been officially involved in the reflection process on Centres of Expertise (CE) for Rare Diseases (RDs) and on European Reference Networks (ERNs) of Centres of Expertise since the beginning of the discussions at EU level.

Without repeating what has already been said in different fora, EURORDIS wishes to contribute to the debate currently going on at both national and European levels aimed at elaborating and establishing an optimal, sustainable system of ERNs throughout the European Union. Through this Position Paper, EURORDIS wishes to underline the main elements from the rare disease patients' perspective for the **implementation phase** of the strategic policy and framework that has been developed in recent years.

EURORDIS is in favour of a step-wise approach aimed at establishing a progression of the activities, recognising that this networking is a process which has to take place mainly in the following fields:

1. Networking in Clinical Care:

ERNs must provide expertise based on Good Practice Guidelines on diagnostic and medical care and on Good Practice Guidelines on Social care, experts' opinion, training, tele-expertise and telemedicine.

2. Information and networking of patients and families:

In this area, networking activities will involve both patients groups and heath care professionals, exchange of experience, information and participation; these activities will be linked to the priorities of the ERNs.

3. Clinical Research:

Activities in this field will include disease registries, research on protocols (compare several treatment options or management strategies), clinical trials (medicines, medical devices, food supplements, etc).

4. Adapted Social Care:

The ERNs will organise respite care, "habilitation/rehabilitation" services, management of social support for complex rare disease cases.

- ➔ The combination of activities to be performed within the ERNs is aimed at establishing comprehensive networks bearing in mind that what rare diseases patients and their families mainly and necessarily expect from the system of ERNs to be established at EU level is reflected in the following recommendations:

1. Overall philosophy of the system:

1.1 **To refuse the logic of making priorities among RDs**: making priorities would result in some RDs being covered, others remaining uncovered thereby creating totally orphaned and abandoned RD patients. EURORDIS therefore recommends that all rare disease patients should be covered by at least one ERN: no patient should be left outside the system, namely each patient should find a "home" in the system.

1.2 **To support a limited number of ERNs** (20 to 30): each ERN would cover a broad range of diseases, gathered by therapeutic areas rather than by distinct disease or limited group of diseases, creating coherent clusters of RDs; e.g. there would be a ERN on "rare pulmonary diseases" instead of one ERN on cystic fibrosis; a ERN on "rare dermatological diseases" instead of one ERN on Epidermolysis Bullosa, etc. The Commission will not be able, even with an increased budget in the 3rd Public Health programme 2014-2020, to support a big number of ERNs for RDs. It has to be kept in mind that ERNs are intended for different health areas, rare diseases being "only" one of them. Therefore, even if RDs have been identified as a priority area for ERNs, EURORDIS suggests to approach all RDs as a whole, and to recommend the creation of a limited number of ERNs for RDs, each Network being supported with significant funding.

1.3 To ensure that ERNs will deliver **structured health care pathways** in a field where relevant knowledge is scattered and patients are not treated following the same standards in all EU countries, thus creating increased inequalities into an area of already great inequity and isolation. These health care pathways will be shared and disseminated throughout the EU.

2. Important characteristics of ERNs must be:

2.1 To ensure a high level of **multi-disciplinarity** within the ERNs, with a large spectrum of medical and scientific competences but also including social aspects. Medical care and social care must be approached simultaneously and on an equal footing in order to succeed improving patients' quality of life. ERNs, as a health care pathway, should cover the whole patient's journey from diagnosis to treatment, including acute events (liaising with the closest points of care and/or the most relevant experts in the network) and re-evaluation of the treatment at pivotal moments of the development of the disease, from high level expertise to continuation of care close to home (proximity care).

2.2 To progressively achieve **coverage of all Rare Diseases**: in a step wise approach between 2014 and 2020 aiming - at the end of the policy process - to cover all rare diseases, as well as **coverage of all EU Member States**. In this way, all EU citizens affected will be covered throughout the European Union.

2.3 To keep a high level of **flexibility**: the structure to be put in place should remain flexible and evolving over time. This flexibility should apply also to the possibility of including external input, when needed, and to the possibility for a patient to turn to a different ERN if needed; e.g. it can happen that at times a patient "belonging" to the ERN on rare pulmonary diseases have to turn to the ERN where expertise is provided mainly in the field of digestive issues. Experts will also have to be mobile to provide this kind of cross-network expertise.

2.4 To guarantee a high level of **inter-operability** certainly between the Networks but also between different departments within the same Network, in order to optimise the level of multi-disciplinarity and flexibility.

3. Involvement in the management, functioning and evaluation of the ERN:

3.1 To recommend that - in addition to **designated Centres of Expertise** recognised at national level - also **patient groups, individual experts, healthcare providers, diagnostic laboratories, social care providers**, be part of the system of ERNs. This is a key outcome of the national conferences on national plans and a key lesson learned from the existing pilot ERNs.

3.2 To underline that patient organisations and **patient's representatives are equal and full partners** in the management and evaluation of the ERNs. Patient organisations (POs) will participate in the governance of the Networks and be members of the Steering Committees/Executive Committees of the ERNs. The EURORDIS Council of European Federations and Networks has elaborated a description of core common tasks and optional tasks to enhance the networking of POs in the relevant therapeutic fields of a ERN while supporting the concrete involvement of POs into key relevant activities, such as biobanking, registries, clinical trials, clinical protocol research, standard on diagnosis & care, standard on social care.

➔ The participation of relevant patient organisations should be one of the criteria of eligibility for funding.

4. Important additional elements:

4.1 **Training** should be provided within the ERNs to health care professionals in order to ensure that professionals are up-to-date in an area of high technology expertise and continuous innovation, including e-health technology. This on-going training will raise the overall level of competence in the field of RDs throughout Europe.

4.2 ERNs must gather a **critical mass of patients and data** in order to support research and improve research capacities. The proposed structuring into a limited number of ERNs (20 to 30) will allow this critical mass to exist and will facilitate interoperability of health information systems across the EU.

4.3 The system of ERNs which will be created should aim at reducing drastically the number of **undiagnosed patients**: a transversal ERN for undiagnosed patients could be organised across the system in order to provide undiagnosed patients with rational pathway toward accurate diagnosis.

4.4 The strategy on ERNs should be carried out with a view at combining it with the development of **National Plans** on RD and the implementation of the **cross-border healthcare** Directive.

4.5 The 3rd PH Programme of DG SANCO (2014-2020) should specifically provide adequate **funding** for the creation and development of ERNs for RD, in a sustainable manner.