

## European Reference Networks for rare diseases: the vision of patients

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Dear Sir,

The European Organisation for Rare Diseases -EURORDIS- is a non-governmental patient-driven alliance of patient organisations and individuals active in the field of rare diseases. EURORDIS represents over 600 rare disease organisations in 58 countries (including 26 EU Member States) covering more than 4,000 rare diseases. More information is available on [www.eurordis.org](http://www.eurordis.org)<sup>1</sup>.

Since 2004, EURORDIS has been involved in different discussions at European and national levels on shaping Centres of Expertise and European Reference Networks (ERNs) for rare diseases with a view to improve timely access to appropriate diagnosis and care for people living with a rare disease.

Over almost a decade, EURORDIS has gathered the opinions of its members on these topics through its Annual Membership Meetings as well as through surveys and workshops organised within the European co-funded projects RAPSODY and POLKA<sup>2</sup>. The opinions of our members have informed relevant EURORDIS position papers, which thus reflect the expectations of patients and their families regarding the organisation of care and delivery of treatments for their diseases.

Why are ERNs important for patients with rare diseases? Due to the low prevalence of each rare disease, expertise is by essence scarce and scattered throughout Europe. The best way to accelerate access to a proper, timely diagnosis, as well as appropriate care, is to share expertise by linking at European level the national Centres of Expertise, healthcare providers, diagnostic laboratories, other relevant services and patients' organisations. ERNs are also intended to gather a critical mass of patients and data to support rare disease registries and clinical research.

The national Centres of Expertise are seen to constitute the nodes of ERNs for rare diseases. EURORDIS, through its representatives, contributed to the Recommendation of the European Committee of Experts on Rare Diseases (EUCERD) on the "quality criteria for Centres of Expertise for Rare Diseases in Member States", which was adopted on 24 October 2011. To date, the EU Member States are developing criteria based on this Recommendation for designated Centres of Expertise for RDs in their territory within their national plans/strategies for rare diseases.

The European Directive 2011/24/EU<sup>3</sup> on the "application of patients' rights in cross-border healthcare" has delineated for the first time the legal framework for European reference networks. This has been a success in translating a concept into a real healthcare framework with an added value for patients.

In addition, Article 13 of this Directive is specifically dedicated to rare diseases: "The Commission shall support Member States in cooperating in the development of diagnosis and treatment capacity in particular by aiming to make health professionals aware of the tools available to them at Union level to assist them in the correct diagnosis of rare diseases, in particular the Orphanet database, and the European Reference Networks".

In this evolving context, EURORDIS adopted in May 2012 a position to outline its vision and strategy for ERNs for rare diseases; what they should comprise and how they should function based on the patient's real life experience.

EURORDIS rejects the logic of selecting priorities amongst rare diseases; all patients affected with a rare disease should be covered by at least one ERN in the long run. As it will not be possible to fund numerous ERNs, EURORDIS recommends that a limited number of ERNs for RDs (20 to 30) should be created by diagnostic and therapeutic areas in order to cover a wide range of rare diseases. EURORDIS also emphasises that a high-level of multi-disciplinarity and inter-operability among ERNs are absolutely necessary. The successful development of ERNs for RDs will rely on a step-wise approach aimed at establishing progressively the various activities of the ERN, such as experts' opinions, good practice guidelines on medical care as well as social care, training, patient registries, e-health. The ERNs should remain flexible in order to evolve over time.

The EURORDIS representatives in the EUCERD voiced the patients' perspectives during the consultations on the elaboration of the Recommendation on European Reference Networks for Rare Diseases. This EUCERD Recommendation<sup>4</sup> was adopted on 31 January 2013 and integrates many elements of the rare disease patients' position.

The new Health for Growth Programme of the European Commission, adopted in March, shall cover a period of seven years, from 2014 to 2020. We are aware that there will not be enough funding for European

reference networks. Therefore, we need to be creative and seek other sources of funding, for instance from the Horizon 2020 programme and possibly structural funds for health. Tonio Borg, European Commissioner for Health, has recently emphasised that research on rare diseases is a priority for the European Commission and where there is an opportunity to offer a better service to citizens, the principle of subsidiarity applies; hence this principle applies to Cross-Border Healthcare for Rare Diseases.

*The Authors declare no conflicts of interest.*

### References

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