



RARE DISEASE PATIENT REGISTRIES



Rare Disease Patient Registries represent a fundamental research effort upon which a number of critical activities are based. They constitute key instruments for increasing knowledge on Rare Diseases (RD) by pooling data for fundamental and clinical research, epidemiological research, and real-life post-marketing observational studies¹. They broadly support health and social service planning by playing a pivotal role in healthcare organisation. They also represent a necessary infrastructure for the implementation of the European Reference Networks for rare diseases, and as such they represent a top priority² for the RD community at a National, European and International level. Furthermore, Patient Registries are one of the main pillars of the current EU policy framework on National Plans for RD³. EURORDIS holds Patient Registries as an advocacy priority and is actively participating in the major EU projects^{4, 5, 6}. In the field, shaping and implementing an EU coordinated strategy on registries that will be patient-centred.

WHY ARE REGISTRIES SO IMPORTANT FOR RD PATIENTS?

It has been demonstrated that Patient Registries are a major determinant for successful translational research in the field of RD. Where well-implemented registries and active patient organizations exist, the likelihood for developing a treatment for the disease in question is increased⁷. Furthermore, the consistent longitudinal collection of patient data facilitates the creation of standards of care and dramatically improves patient outcomes and life expectancy even in the absence of new therapies. These compelling arguments for Rare Disease Patient Registries as indispensable infrastructure tools for translating basic and clinical research into therapeutic solutions have elevated their status to a major priority for all stakeholders, making them a building block of any sound policy on RD.

HOW TO ADDRESS THE ISSUE?

Being very thorough research infrastructures, Patient Registries require significant time and human resources as well as conspicuous financial investment and sustainability plans. Standard operating procedures and common resources or platforms for centralizing new or existing registries should be developed. Collaborative efforts at all levels are paramount to establish and manage Rare Disease Patient Registries and derive from them meaningful outcomes in the most efficient manner and for the uppermost benefit of patients. Nevertheless, no uniform, accepted standards currently govern the collection, organisation, or availability of data collected by Rare Disease Patient Registries. The existence of conspicuous data-sharing barriers creates a compelling argument for developing globally accepted definitions, classifications, data standards and favourable and congruent policies and resources facilitating data sharing and pooling.

DEVELOPING RARE DISEASE PATIENT REGISTRIES: THE 10 MAJOR PRINCIPLES PROMOTED BY EURORDIS²

- **Patient Registries should be recognised as a global priority in the field of Rare Diseases.**
The priority for establishing and supporting them has become more and more compelling and urgent for all stakeholders in order to achieve their different but complementary goals aimed at augmenting knowledge and developing new therapeutic breakthrough in the field of RD.
- **Rare Disease Patient Registries should encompass the widest geographic scope possible.** The benefits of international collaboration and maximization of limited resources are most obvious for rare diseases, especially the very rare ones, due to the low individual prevalence and the scarcity of information related to each of them.
- **Rare Disease Patient Registries should be centred on a disease or group of diseases rather than a therapeutic intervention.** Disease focused registries ensure higher quality and completeness of data and allow fulfilling post-marketing requirements, especially for regulators and payers, assessing the long-term outcomes and cost-effectiveness of new medicinal products.
- **Interoperability and harmonization between Rare Disease Patient Registries should be consistently pursued.** It has become a compelling necessity to develop globally accepted definitions, classifications, data standards and favourable and congruent policies and resources facilitating data sharing and pooling. Ideally, standard operating procedures and common resources or platforms for centralizing new or existing registries should be developed.
- **A minimum set of Common Data Elements (CDE) should be consistently used in all Rare Disease Patient Registries.** This would facilitate the standardization of data (same definitions and entry procedures), harmonization (facilitation of data exchange and comparison), and interoperability (shared quality assurance and data security standards).

- **Rare Disease Patient Registries data should be linked with corresponding biobanks data.** The high value of RD biological samples only increases when coupled with well-documented, associated data housed in a patient registry. The development of a system that assigns a unique global identifier to each patient is recommended to facilitate data linkage and avoid duplicate entries and waste of precious biomaterial.
- **Rare Disease Patient Registries should include data directly reported by patients along with data reported by healthcare professionals.** By complementing clinician-reported data in Rare Disease Patient Registries, patients can contribute to improving their robustness, comprehensiveness and quality. Continued creation of easily accessible and validated standards, platforms and scientific guidance to ensure the high quality collection of patient-entered data should be encouraged and guaranteed.
- **Public-Private Partnerships⁸ should be encouraged to ensure sustainability of Rare Disease Patient Registries.** The development of each individual registry or network of registries requires significant effort and financial commitment. With both governments and private groups showing interest in patient registries, public-private partnerships are a promising collaborative scheme.
- **Patients should be equally involved with other stakeholders in the governance of Rare Disease Patient Registries.** Patients should be involved at all levels of development, management and maintenance in order to best represent patient needs, increase awareness among all stakeholders of the existence of the registry and, ultimately, improve the quality and quantity of data collected.
- **Rare Disease Patient Registries should serve as key instruments for building and empowering patient communities.** The creation of a patient registry can facilitate the getting together of patients and their families as they engage directly into the development of the very databases in which their data will be entered. Registries thus become the medical home for patients scattered internationally and empower patients with data available for sharing with health care professionals, clinical researchers and drug developers.

EU POLICY FRAMEWORK ON RARE DISEASE REGISTRIES

- **The Communication from the Commission, "Rare Diseases: Europe's Challenges"⁹** proposes that Member States (MS) put in place strategies aimed at ensuring mechanisms to gather national data on RD and pool it together with European counterparts.
- **The Council Recommendation on an Action in the Field of Rare Diseases¹⁰** accompanying this communication further highlights the importance for the development of research and healthcare infrastructures in the field of RD. Patient registries can be listed as such infrastructures and given their long-lasting nature, the need for appropriate financial provisions at the national level to ensure their sustainability is vital.
- **The European Project for Rare Diseases National Plans Development (EUROPLAN) recommendations³** underlines the importance for MS to stimulate and support national initiatives in a European or international framework in the domain of registries and of their uses for research, epidemiology and clinical purposes, and for health and social services planning.

EU POLICY FRAMEWORK ON RARE DISEASE REGISTRIES

- **The EUCERD (the EU Committee of Experts on Rare Diseases¹¹)** will monitor and finally synthesize the outcomes of the major EU projects on registries (EPIRARE, PARENT and RD-CONNECT^{4,5,6}), working with the European Commission on a strategy for a possible European reference platform for Rare Disease Patient Registries. Also, the EUCERD plans to adopt in the course of 2013 a Recommendation on Databases and Registries for Rare Diseases to guide the shaping of both EU and national policies on the subject.

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