Patients Registries for Rare Diseases Workshop
Madrid, 21-22 March 2017

Workshop Rationale and Goals

The EU has recommended that Member States should consider supporting at all appropriate levels, including the Community level, on the one hand, specific disease information networks and, on the other hand, for epidemiological purposes, registries and databases, whilst being aware of an independent governance. (Council Recommendation on an action in the field of rare disease (2009/C 351/02).

Patient registries and databases constitute key instruments to develop clinical research in the field of rare diseases, to improve patient care and healthcare planning. They are the only way to pool data in order to achieve a sufficient sample size for epidemiological and/or clinical research. They are vital to assess the feasibility of clinical trials, to facilitate the planning of appropriate clinical trials and to support the enrolment of patients as well as for the post-marketing surveillance of orphan medicinal products.

The European Commission has already established a **European Platform on Rare Diseases Registration** which provides common services and tools for the existing (and future) rare diseases registries in the EU. This platform can constitute a powerful tool to create and structure networks of experts, whether they being European Reference Networks of Centres of Expertise or national expert networks for RD. In either case, the experts and centres of expertise involved are a primary source of data for registries.

This workshop will bring together all the stakeholders key for the development of patient registries for rare diseases allowing them to share the results from several EU Health Programme Projects and Joint Actions funded between 2008-2015, fostering the exchange of knowledge and best practices among European experts and countries.

Context: Technical projects on rare diseases registration

The Health Programme is supporting the **EPIRARE (European Platform for Rare Disease Registries) Project**, in order to build consensus and synergies to address regulatory, ethical and technical issues associated with the set up and management of registries for Rare Diseases patients in the EU and to contribute to prepare a platform for the registration of rare disease patients in Europe and to ensure the quality and best use of the registered data.

The aim of the **PARENT Joint Action (Cross Border PATient REgistries iNiTiative)**, under the Health Programme, is to support MS in developing comparable and coherent patient registries in fields where this need has been identified (e.g. chronic diseases, rare diseases, medical technology), and to support MS states in the provision of objective, reliable, timely, transparent, comparable and transferable information on the relative efficacy and effectiveness of health technologies.

The **FP7 Project RD-CONNECT (An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research)** will provide an integrated, user-friendly RD-Connect platform, built on efficient informatics concepts already implemented in international research infrastructures for large-scale data management, will provide access to federated
databases/patient registries, biobank catalogues, harmonised -omics profiles and cutting-edge bioinformatics tools for data analysis.

Objectives of IRDiRC (International Rare Diseases Research Consortium) in the field of rare diseases registration, in a transatlantic basis, are in the direction of a meta-registries or registry of registries as suggested by the agency for Healthcare Research and Quality (AHRQ USA). A registry of registries should prove to be very helpful to the public who are seeking an appropriate patient registry for patient participation.

The EUCERD (European Union Committee of Experts on Rare Diseases) adopted on 5th June 2013 the following recommendation: EUCERD Core Recommendations on Rare Disease patient registration and data collection.

Aim of the Workshop:

The aim of the conference is to present results of the EU Health programme projects funded between 2008-2015 (EPIRARE Project, PARENT Joint Action, E-HOD, E-IMD, etc.) in order to achieve the following specific objectives:

- To foster knowledge exchange and best practices of these European registries.
- To reinforce research and epidemiological surveillance, through setting up of shared registries, among others.
- To maximise integration and interoperability of EU and national rare disease registries.
- To highlight those registries linked to approved ERNs
- To provide a forum to discuss the present and future situation of patient registries.

The target audience to be reached includes health professionals, academia, health policy makers and civil society organisations.