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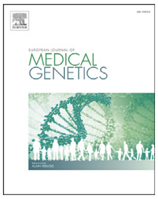

How the EUCERD Joint Action supported initiatives on Rare Diseases

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ABSTRACT

Joint Actions are successful initiatives from the European Commission (EC) that have helped to raise awareness and to bring significant benefit to those suffering from a rare disease (RD). In this paper, we will focus on the activities developed by the EUCERD Joint Action (EJA) and by the Orphanet Joint Action ("Orphanet Europe"). EUCERD Joint Action was co-funded by the EC and the Member States between 2012 and 2015 to help to define the activities and policies in the field of RD and foster exchange of experiences amongst Member States. This project is the continuation of previous efforts to turn RD a priority in the EC Health Programmes. "Orphanet Europe" was a Joint Action co-funded by INSERM, the French Directorate General for Health and the EC to address the need for a common portal that would gather the most update information regarding RD. This need was identified in the European Commission report "Rare Diseases: Europe's challenge" and in the Recommendation of the Council for a European RD portal. These joint actions have supported the policy development work of the European Commission, through the support of their committees for rare diseases. In this paper, the authors aim to raise awareness of the work done by the EUCERD Joint Action on behalf of the rare disease community and the policies established.

Contents

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1. Introduction

Joint Actions for rare diseases (RD) are collaborative initiatives co-funded between the EU and Member States that have had a clear EU added value helping with the development and dissemination of RD policies across all Member States. The Joint Actions in the field of RD (EUCERD Joint Action (EJA) and Orphanet Europe) have given technical support to the work developed by the EU Committee of Experts on Rare Diseases (EUCERD) and the Commission Expert Group on Rare Diseases (CEGRD).[1]

With an estimated population of approximately 30 million, RD have a major socio-economic impact on European social and medical systems. Individual RD are still not well captured in health information systems (Aymé et al., 2015), and the true economic burden of the diseases can be difficult to demonstrate in spite of the different RD dedicated programmes established by the EC.

The ideals for a necessary common approach to RD in the different Member States are encompassed in the Commission Communication (Commission Communication, Rare Diseases Europe's Challenge, 2008) and in the Council Recommendation (Council Recommendation on an action in the field of rare diseases, 2009). These documents define a series of priority areas where action should be focused so that the recognition and visibility of RD could be raised. One of the major goals of the EC regarding RD was to help Member States to develop coherent National policies (National Plans/Strategies for RD) and to stimulate international cooperation and coordination regarding aspects such as coding, classification, definition of Expert Centres or genetic testing amongst others.

The Council Recommendation focuses on several key themes: the definition, codification and inventorying of RD; research; European Reference Networks (ERNs); gathering expertise at EU level; empowerment of patient organisations; and sustainability of health policies for RD. Work done by different stakeholders allowed the identification of the major constraints and possible solution to tackle the RD challenge. It became consensual that when the "*numbers*" are low the best way to progress is through networking and sharing of expertise and data.

2. Joint Actions for Rare Diseases and their role on policy development

2.1. The EJA and EU Committee of Experts on Rare Diseases

The RD field has so many specificities; that go from small number of patients with one disease to the small number of experts and the dispersion of resources; that the actions taken to tackle these diseases need to be addressed, first by the existing experts without omitting the patient representatives. Only after a profound analysis and discussion can actions in the field of RD be transformed in policies.

Considering the need to have advice from RD experts, in 2009, an EU Committee of Experts on Rare Diseases (EUCERD) was convened to support the European Commission in formulating and implementing RD policy. This group expanded the work initially overseen by the Rare Disease Task Force (RDTF), established in 2004 (Aymé and Rodwell, 2014). In 2013, the EUCERD was reformed as a Commission Expert Group on Rare Diseases (CEGRD). The remit of this multi-stakeholder group (all Member States are represented, as well as patient organisations, industry, learned societies and experts) is to assist the European Commission in drawing up legal instruments and policy documents through the preparation of guidelines, opinions and recommendations. Table 1 shows the documents submitted to the EC following approval of the EUCERD or CEGRD expert groups (Table 1). The documents were first prepared and researched by the EUCERD Joint Action members, following wide-ranging consultation and input from experts and opinion leaders, which generated these policy documents, and submitted to the Expert Group for discussion and final approval.

2.2. EUCERD Joint Action (EJA)

The EUCERD Joint Action (EJA) was funded from 2012 to 2015 to continue the work previously undertaken by the Rare Disease Task Force, the EUROPLAN project, and the various rare disease networks that had received EU funding in previous EU programmes (FP6, FP7, Public Health Programme, etc.). The EJA focused on a broad range of topics, in agreement with the EC, across the RD policy area and supported the work of the committees in the generation and review of recommendations (Table 1). The EJA also ensured the integration of work with other relevant activities such as parallel joint actions and research projects, such as the Orphanet Joint Action and RARECAREnet (rare cancers). The primary aim of the EJA was to support the work needed to implement the Council Recommendation and the Commission Communication on RD.

3. State of the Art of Rare Disease Activities in Europe

A major focus of the EJA was on mapping and enhancing countries' efforts to structure RD activities within the health and social systems. The annual report on the *State of the Art of Rare Disease Activities in Europe*, produced as part of the EJA, and continuing in RD-ACTION (see below), gave maximum visibility to pan-European (and global) RD developments based on data provided by Orphanet and from Member State consultation. This report covered the political framework, expert services, research and development, orphan drugs and information services and with over 15,000 downloads per year is an invaluable resource for information on RD activities across the Member States (Rodwell and Aymé, 2014).

4. National plans/strategies for rare diseases

The Member States while signing the Council Recommendation on an action in the field of RD, committed themselves to elaborate and adopt National Plans/Strategies for RD by 2013. To build awareness and capacity at the country level, and to support the elaboration of national plans/strategies for RD the EJA helped to fund and deliver 21 national conferences in EU Member States (and supported the organisation of similar events in Georgia, Russia, Serbia, and Ukraine). The organisation of these national conferences by EURORDIS and national patient alliances, with the participation of the National health authorities, helped to assess the applicability and commitment of EU policies at a national level. By September 2015, 24 countries had published a national plan or strategy for RD, with some already working on a second plan.

The EJA helped to establish a network of stakeholders, mainly policy makers and patient representatives, which was able to share relevant national experiences and in this way contribute to the goal of having equity in care for RD patients.

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[1] http://ec.europa.eu/chafea/documents/health/leaflet/chafea-joint-action-2015_en.pdf.

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