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Policy recommendations for rare disease centres of expertise

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ABSTRACT

Aim: Rare diseases are a serious public health concern and are a priority in the EU. This study aims to develop policy recommendations for rare disease centres of expertise (CoEs) in order to improve standards and quality of care.
Subject and methods: A modified 3- round Delphi technique was used. Participants included rare diseases patients, carers, patient representatives and healthcare professionals (HCPs) from CoEs in two countries—Denmark and the UK.
Results: The results suggest the need to make improvements within current CoE environments, access to CoEs and the need for coordination and cooperation of services within and outside CoEs. It is recommended that CoEs are not overly 'medicalised', while at the same time they should be established as research facilities. The importance of including patient representatives in CoE performance management was also highlighted. Raising awareness and provision of appropriate training amongst non-specialist HCPs is seen as a priority for early and correct diagnosis and ensuring high quality care. Similarly, provision of targeted information about patients' illness and care was considered essential along with access to social assistance within CoEs.
Conclusions: Policy recommendations were developed in areas previously recognised as having gaps. Their implementation is expected to strengthen and improve current care provision for rare disease patients. In member states where national plans and strategies are being developed, it is recommended to replicate the methodological approach used in this study as it has proven to be a helpful tool in rare disease centres of expertise policy development.

1. Introduction

A rare disease (RD), also referred to as an orphan disease, is any disease with a particularly low prevalence. In the European Union (EU) a disease is classified as an RD when it affects less than 5 persons per 10,000 (EU CERD, 2011) on a population level. It is estimated that between 5000 and 8000 distinct RDs exist, affecting a total of between 6% and 8% of the EU population over the course of their lives (EU CERD, 2011). Although rare diseases are characterised by a low prevalence for each of them individually, the total number of people with a rare disease is estimated in the EU to be between 27 and 36 million (EU CERD, 2011).

Rare diseases are a serious public health concern and are a priority in the EU (European Commission). Currently, in European member states (MS) and at a European level there is a need for robust strategies, plans and specific policies on rare diseases (European Commission, 2008). Few European countries have specialised provision of health services for rare diseases. Despite a significant EU population being affected by RDs healthcare systems in MS are not set up adequately to provide care for RD patients. In many European Union (EU) Member States (MS), RD patients are subject to marginalisation in classic healthcare systems designed for non-rare diseases (Kole & le Cam, 2010). As a result, they do not experience equal access to timely, high quality health services they deserve (Kole & le Cam, 2010). The restructuring of healthcare systems to better reflect the values of equity and solidarity amongst RD patients, professionals, and policy makers across Europe needs to be accomplished.

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EU health ministers recognise the challenges and need for action in the field of rare diseases which has led to developing a set of recommendations. The development of Centres of Expertise (CoEs) and European Reference Networks (ERNs) in the field of RD is encouraged in the 'Council Recommendation on an Action in the Field of RD (2009/C 151/02) (8 June 2009)' and most recently in the 'Directive on the application of patients' rights in cross-border healthcare (2011/24/EU) (9 March 2011)' as a means of organising care for thousands of heterogeneous RD affecting scattered patient populations across Europe (Rodwell, Aymé, & Bushby, 2012). The recommendations concentrate on supporting and strengthening EU countries' plans and strategies for responding to rare diseases, improving recognition and visibility of rare diseases, encouraging more research into rare diseases and forging links between medical centres. No official definition of a RD CoE exists, however ideally they are medical centres that bring together a group of multidisciplinary, specialised competencies and ensure timely diagnosis and appropriate follow-up care by aiming to improve the continuity and coordination of care through the implementation of healthcare pathways and research (Kole & le Cam, 2010). A ERN is considered to be a physical or virtual networking of national RD CoEs to exchange knowledge and expertise at a European level.

As part of the EC's commitment to improving healthcare for rare disease patients, in 2008 a 3-year project called 'POLKA: Patients' consensus on preferred policy scenarii for rare diseases' was funded and supported by the European Commission DG Sanco. The main objective of the POLKA project was to facilitate the consultation of the European rare disease community, with the aim of building consensus on preferred public health policy scenarios for rare diseases, including the quality of care available at RD CoEs, and if that level of quality was sufficient.

In order to achieve the POLKA project's objective of gathering expert advice and experience and building consensus on policies for Centres of Expertise on rare diseases, a study within POLKA called 'The POLKA Delphi study' was undertaken. This article presents the results of the POLKA Delphi study and the policy recommendations developed from them aimed at better provision of healthcare for RD patients in Europe.

2. Aims

The aim of the POLKA Delphi study was to:

- Assess experiences of healthcare professionals (HCPs) and patients (including patient representatives and carers) at selected CoEs,
- identify policy areas with gaps for discussions on the current services and,
- develop policy recommendations based on findings in order to help improve care for rare disease patients across Europe.

3. Methods

3.1. The Delphi technique

A 'Delphi technique' is defined as "a method for structuring a group communication process so that the process is effective in allowing a group of individuals, as a whole to deal with a complex problem." (Linstone & Turoff, 1975). It is based on the premise that 'pooled intelligences' which enhances individual judgment and captures the collective opinions of experts (Linstone & Turoff, 1975; Moore, 1987; Murry & Hammons, 1995).

There are several types of Delphi. Based on a classification by van Zolingen and Klaassen (2003) they are the Classic Delphi, the Policy Delphi, the Decision Delphi and the Group Delphi. The different forms of the Delphi such as the Policy, Decision and Group Delphi are variations of the classical Delphi. There are many more variations of Delphi now in existence known as 'modified Delphi' (McKenna, 1994) as researchers modify the Classic Delphi technique to suit their needs and few researchers nowadays see the need to use uniform methods. In order to meet the aims of the POLKA Delphi study a modified-Policy Delphi technique was employed.

3.2. The POLKA Delphi technique

The POLKA Delphi study was planned to be carried out in RD CoEs from three countries—Denmark, the UK and France. The countries were chosen as their healthcare systems were amongst the most developed ones set up to provide specialised care for patients with rare diseases in Europe. In majority of the MS, there is a lack of specialised services for patients with rare diseases through specialist healthcare centres/CoEs. This paper presents results from Denmark and the UK as the French arm of the study encountered significant issues recruiting healthcare professionals. Fig. 1 illustrates the stages of the study reported here. The Delphi process consisted of 3 rounds. The first step of the Delphi process was the selection of participants for the Delphi panel followed by administering a questionnaire survey (Round 1). The results of the questionnaire survey were fed back to the participants and used to facilitate discussions at face-to-face meetings (Round 2). The results of both rounds were collated and used to develop policy recommendations. The draft policy recommendations were sent to participants for validation and sign-off (Round 3).

3.3. Participants

A total of eight CoEs[1], one in Denmark and seven in the UK, participated in the study. A contact person at each of the eight CoEs was identified to recruit a total of 160 participants as the Delphi panel and coordinate the study – 8 HCPs and 12 patients (or patient representatives and carers) per CoE – see Table 1.

3.4. Round 1: Questionnaire survey

Two questionnaires, one for HCPs and one for patients, were developed by the authors (AMS & ARA) in collaboration with the POLKA project members with the aim of assessing the current experiences of healthcare professionals and patients at the centres of expertise. The questionnaires were based on the results of a survey called 'Eurordiscare3'[2] which gathered data on patients' experience and expectations concerning health services for rare diseases in Europe.

The majority of the questions asked in the HCP and patient questionnaires were similar except for a few which were specific to each group. The questionnaire for HCPs consisted of 46 questions and the questionnaire for patients consisted of 39 questions. Questions in both HCP and patients questionnaires were divided into the following 13 policy areas:

(i) Environment at CoEs,
(ii) access to CoEs,
(iii) coordination and cooperation within and outside CoEs,
(iv) transition of care (childhood to adulthood),
(v) patient autonomy,

[1] It should be noted that the CoEs are currently not 'labelled' as CoE and but are specialised health services provided at hospitals for rare disease patients.

[2] Available at http://www.eurordis.org/IMG/pdf/voice_12000_patients/EURORDISCARE_FULLBOOKr.pdf.