



**RB-REP.02**

## **Publishable Summary**

*01 January 2013 – 30 June 2014*

**Platform for sharing best  
practices for management of  
rare diseases**

***(RARE-Bestpractices)***

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## 1. Summary description of project context and objectives

Rare diseases (RD) are characterized by low prevalence and extreme diversity as regard origin and clinical manifestations. Many RD are life-threatening or fatal, most of them are chronic and seriously debilitating. Collectively they affect millions of people of all ages worldwide.

The small number of people affected with any particular RD as well as the large number of RD result in challenges which complicate research progress and knowledge creation in all countries. These challenges include difficulties in attracting public and private funding for basic and clinical research, in setting up appropriate studies for translating research findings into practice, in coordinating research initiatives. For patients this means late diagnosis, as well as late, inadequate or even harmful treatment, and low quality of care.

RARE-Bestpractices intends to address such international critical challenges improving the management of RD patients and promoting a consistent level of healthcare services in European countries. RARE-Bestpractices aims at creating a sustainable networking platform offering high quality information to health professionals and to patients and their carers, and facilitating communication in the field of guideline development, health technology assessment, and horizon scanning procedures.

The objectives of RARE-Bestpractices are as follows:

- reach consensus on the methodology to develop guidelines on RD
- build a comprehensive public database of trustworthy guidelines to help professionals, patients, policy makers with the best and most up-to-date information on RD
- build a comprehensive public database of research recommendations to identify and prioritize RD research needs
- define to what extent conclusions from cost-effectiveness analyses for pharmaceuticals are accounted for and implemented in best practice guidelines across a range of countries
- set up training activities targeted at key stakeholders to spread expertise and knowledge in the field of guidelines on RD
- support IRDiRC (International Rare Diseases Research Consortium, <http://www.irdirc.eu>) activities as regard both the translation of research results into patient-oriented strategies and the identification of research needs.

The project brings together a team of experts in the area of guidelines, systematic reviews, health technology assessment, health policy, RD epidemiology and public health coming from 9 countries across Europe.

The work has been organized into eight work packages (WP) to allow an efficient coordination of the project activities. WP3, WP4 and WP5 include the core research activities, dealing respectively with setting the quality standards for the development of RD guidelines, collecting existing guidelines and research recommendations on RD, evaluating orphan drug appraisals to explore processes and best practices in a range of countries. WP6 aims at building awareness around the project and disseminate the results. Resources of WP6 are also dedicated to the foundation of new open access journal which promotes analysis and discussion on improving health and health care

on rare diseases. WP2 supports WP3, WP4, WP5 and WP6 activities and outputs by developing the platform technical infrastructure. WP7 is responsible for establishing collaboration with the International Rare Diseases Research Consortium (IRDiRC). WP1 (scientific coordination) and WP8 (management) focus on ensuring a smooth execution of project activities.

## 2. Work performed since the beginning of the project and main achieved results

During the first period of operation (January 2013 – June 2014) major effort was put into the **successful launch of the project** and the establishment of a sound basis **for a fruitful cooperation** of the project partners towards the research objectives. This has been achieved by effective leadership (WP1) and project management (WP8).

In WP1, work has also been done on planning a series of **training events** on guideline development and evaluation.

For this first 18 months reporting period WP3 was focused on exploring challenges in developing recommendations included in RD guidelines. Two workshops have been organized to explore issues specific to RD and to assess the use of the GRADE approach in creating RD guidelines. Consortium participants, patient representatives, invited experts in the field and external advisors (e.g. the GRADE working group) were involved in the process. Results of this work -have been summarized in a paper submitted to a peer-review journal.

The agreed methodology will be applied and tested in the production of **a real guideline** on catastrophic antiphospholipid syndrome. A draft work plan has been defined. Contacts have been established with key experts on this disease, in particular with the American Society of Hematology.

WP4 is in charge of **collecting existing guidelines and research recommendations on RD**. The work focused on scoping searches and developing the collection criteria. The collection methodology has been reported in a peer-reviewed article (*Rare Journal*, Vol. 1, No. 1). Both the collection criteria and the database specifications have been informed by a preliminary information needs analysis through a survey directed to project partners and advisory board members. Patient input were also obtained. AGREE II tool was selected for guideline evaluation.

WP2, responsible for the **technical infrastructure** of the platform, designed and implemented both databases (currently in user testing phase), a forum based web community (prototype ready for testing), and also the infrastructure of the project website [www.rarebestpractices.eu](http://www.rarebestpractices.eu) launched in June 2013.

WP5 is leading the work of mapping out the policies currently in place in eight European countries on **orphan drugs appraisals** and **exploring the implication these policies have for coverage decisions**. A review of the available literature and jurisdiction-specific guidelines on the value assessment of orphan drugs was performed and presented in a peer-reviewed article (*Rare Journal*, Vol.1, No.3). The ongoing research is directed to understand how appraisals are conducted, the critical factors leading to positive and negative recommendations and how do assessments feed into clinical practice guidelines. For this reason, 17 case studies on 17 drug-

indication pairs are being conducted based on appraisal documents and publicly available guidance.

WP6 is in charge of **disseminating the project and its achievements**. Several dissemination tools have been realised e.g. newsletters, leaflet, Twitter account ([@RAREBestP](#)), press releases and a large number of publications. WP6 also created and continuously updates contents for the web site, which was conceived as an efficient and effective publishing environment that suit the needs of all users. A dissemination plan (to be updated annually) encouraged partner involvement optimizing participation. The project was presented in a number of international conferences (e.g. ECRD 2014, IFLA WLIC 2014) and a workshop was organized to present the project to patients at the EURORDIS membership meeting.

Moreover extensive efforts have been made in WP6 for the creation and publication of a **new science open access journal** *Rare Diseases and Orphan Drugs. An International journal of public health* (<http://rarejournal.org>).

Finally, RARE-Bestpractices partners actively participated in all the activities of 2013 of **IRDIRC** providing important input and working to establish a proactive partnership.

### **3. Expected final results and their potential impact and use**

RARE-Bestpractices intends to offer a response to the challenges of improving quality and consistency of best practices for RD, in accordance with the recommendations of the European Union Directive which encourages European member states to provide high-quality and quantitatively adequate healthcare to citizens as well as to produce “good practices guidelines” (article 12. 3,4 - Directive 2011/24/EU).

The value of health care guidelines has also been recognized by the more recent Commission delegated decision (C(2014)1408 f) which includes “develop and implement clinical guidelines” among the criteria and conditions that European Reference Networks must fulfill.

Unfortunately, a multitude of approaches in developing, evaluating, disseminating guidelines are currently utilized across European countries. Moreover a lack of guidelines for the proper management of patients is currently experienced within the RD community.

RARE-Bestpractices will offer a methodology which explores issues specific for RD, assessing the use of GRADE approach in creating guidelines for RD and drawing on existing initiatives in the field of “standards for guidelines”. The enhancement of methodological rigor of health care guidelines will increase the quality of guidelines and ultimately the health care for RD patients in a positive circle.

In addition, RARE-Bestpractices will ensure that the large RD community has access to “trustworthy” guidelines. A comprehensive system of resources facilitating communication and cooperation in the field of guidelines on RD (collection of guidelines, training tools, etc.) will be made publicly available.

The dissemination of the outputs of the project will be maximized by the wide network involved in the project. Experts in RD research area and experts in guidelines and health technology assessment area, brought together for the first time, from academic institutions, agencies, organizations, patient representatives, governmental bodies will ensure uptake of the innovative findings of RARE-Bestpractices by the RD community, including RD national plans, Centres of Expertise and European Reference Networks in compliance with European Union legislation.

## 4. Contact details

For additional information please visit the Project WEB site: [www.rarebestpractices.eu](http://www.rarebestpractices.eu)

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