

# RARE-BestPractices

## Building a model collection of guidelines for rare diseases

Karen Ritchie, Lorna Thompson and Jenny Harbour  
Healthcare Improvement Scotland

<http://www.rarebestpractices.eu/>

### The project

RARE-Bestpractices is a four-year European Union (EU) 7th Framework project which aims to improve the management of patients through the sharing and development of rare disease (RD) best practice, knowledge and information. The project focus is to collect, evaluate and disseminate existing guidance on rare conditions, to cultivate a robust methodology and suite of tools appropriate to RD clinical guideline development, and to facilitate collaboration, knowledge exchange and research in the area of RD.

### The issue

Over 30 million people in Europe have a rare disease. There are approximately 6000 recognised rare conditions, defined as affecting no more than 5 per 10000 persons. Effective clinical management can be impaired by lack of evidence for decision-making, limited treatment options, limitations in access to specialist services and clinicians' lack of knowledge and experience of low prevalence disorders.

### Aims

This work aims to create a database of quality appraised guidelines for rare diseases.

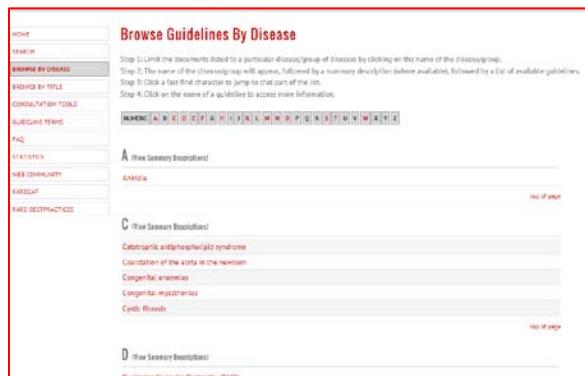


Fig 1. Screenshot of the guidelines database, browse by disease

[www.rbguidelines.eu](http://www.rbguidelines.eu)

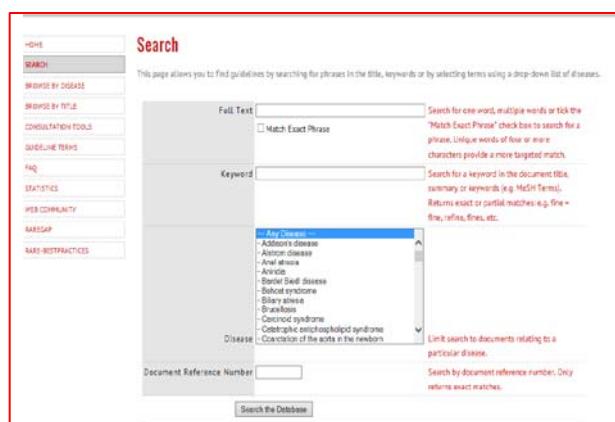


Fig 2. Screenshot of the guidelines database, search facility

### Methods

- A set of 44 rare disease topics for the initial model collection was identified using informal consensus
- A literature search protocol was developed specifically to identify published guidelines on rare disease topics
- Dual appraisal of each guideline using AGREE II was facilitated
- Guidelines and associated information were submitted to the web-based RAREGUIDELINE database

The assessment of the AGREE II instrument as a suitable tool to appraise the methodology of rare disease guidelines was published in Volume 2 of the project journal 'Rare Diseases and Orphan Drugs'

[www.rarejournal.org](http://www.rarejournal.org)

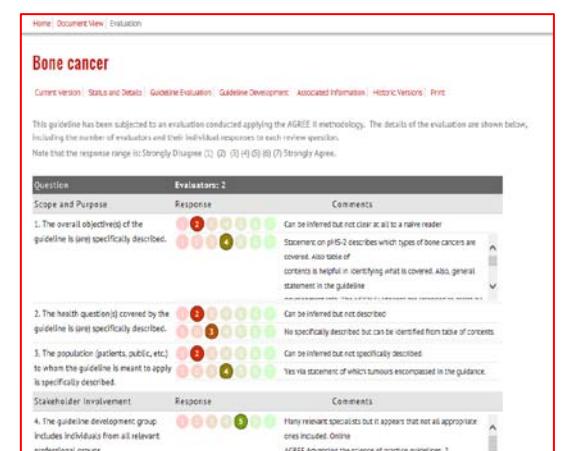


Fig 3. Screenshot showing guideline evaluations and comments.

### Results

Rare disease specialists, patients representatives and guideline developer partners from across Europe have contributed to the development of the collection. To date over 50 guidelines have been identified, appraised and submitted to the web-based model collection.