

## Towards Patient centered care in rare diseases

EAP's experience from RareBestPractice Project  
2013-2016

The Rare best practice project has collected best practice guidelines and identified rare disease research needs. Over 200 rare best practice guidelines are identified and systematically appraised with the AGREE II tool. The ultimate goal of the project is to facilitate timely, effective and efficient translation of research into patient oriented clinical and public health practice.

The results as presented in the meetings in Rome on November 23<sup>th</sup> and 24<sup>th</sup>, illustrate that:

- 1- A substantial number of specific guidelines on rare diseases exist: <http://www.rbpguidelines.eu> ;
- 2- There are still considerable gaps: for example, while there are 24 guidelines on cystic fibrosis none is available on congenital cataract. This is a remarkable fact because most children in Europe are screened for cataract in the first months for an early access to treatment;
- 3- Patient (organisation) involvement is one of the major missing elements in the rare disease guidelines;
- 4- There may be conflicts of interest when a small group of experts define the care they provide as the basis of the guideline, especially when there is no patient (organisation) involvement;
- 5- It is not always clear whether research funding by industry may influence the recommendations in guideline ;
- 6- It is possible to produce a rare disease guideline with the open access GRADE tool;
- 7- The application of guidelines can generate insight in best care for best price with reduction of unnecessary and repeating investigations, and visits to health care providers as well as effectiveness of treatments;
- 8- HTA and Health care delivery can be improved when data and processes for their management, now locked in vendor dependant IT health systems, will satisfy the paradigm of interoperability;
- 9- Healthcare processes, clinical practice guidelines, healthcare pathways can be formalised and represented with the already existing standardised graphical languages (e.g., Unified Modeling Language®, Business Process Modeling and Notation). This allow to avoid logical gaps in the planning phase, and enhance clarity avoiding misinterpretation in the application phase and, finally, to support process enactment in the implementation of information systems. Moreover, ontology languages (preferably worldwide recognised standard such as RDF and/or OWL) should be use in order to represent the knowledge regarding data collection and process interoperability.
- 10- Scientific societies are responsible for the specialist care they provide, patient organisations have become very active to join the health care providers towards a better and affordable health care, once supported by open IT systems.

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*The Convention of the Rights of the Child* and the *Convention of the Rights of Persons with a disability* highlight how children with disabilities have the same rights as other children.