



RARE
DISEASES
INTERNATIONAL

STATE OF THE ART OF RARE DISEASE ACTIVITIES: HOW TO COLLECT AND BENCHMARK RARE DISEASE POLICY INFORMATION

BEYOND THE EU?

Paloma Tejada

Director, Rare Diseases International

RDI Membership Meeting , Vienna

May 10, 2018

THINK ABOUT THESE QUESTIONS

- ▶ How would I use a State of the Art of Rare Diseases to advocate in my country?
- ▶ How could I collect and verify this type of information in my country?



WHAT IS THE STATE OF THE ART?

- ‘State of the art of RD activities in Europe’ report
- Well-established resource providing valuable, detailed information for all stakeholders in the field of RD and orphan medicinal product
- “Go to’ source of information on RD activities in Europe
- It useful to benchmark RD policy throughout EU
- Building block towards an international standard



BACKGROUND TO SOA REPORT

- Conceived in 2011
- Originally funded under EUCERD Joint Action
- 3 editions (2012, 2013, 2014), each one 5 volumes (around 500 pages)
 - Plus individual country reports
- Around 15'000 downloads (all volumes combined) annually
- Content based largely on OrphaNews articles and developments.
- In 2016, the SoA migrated to RD-ACTION and managed by UNEW



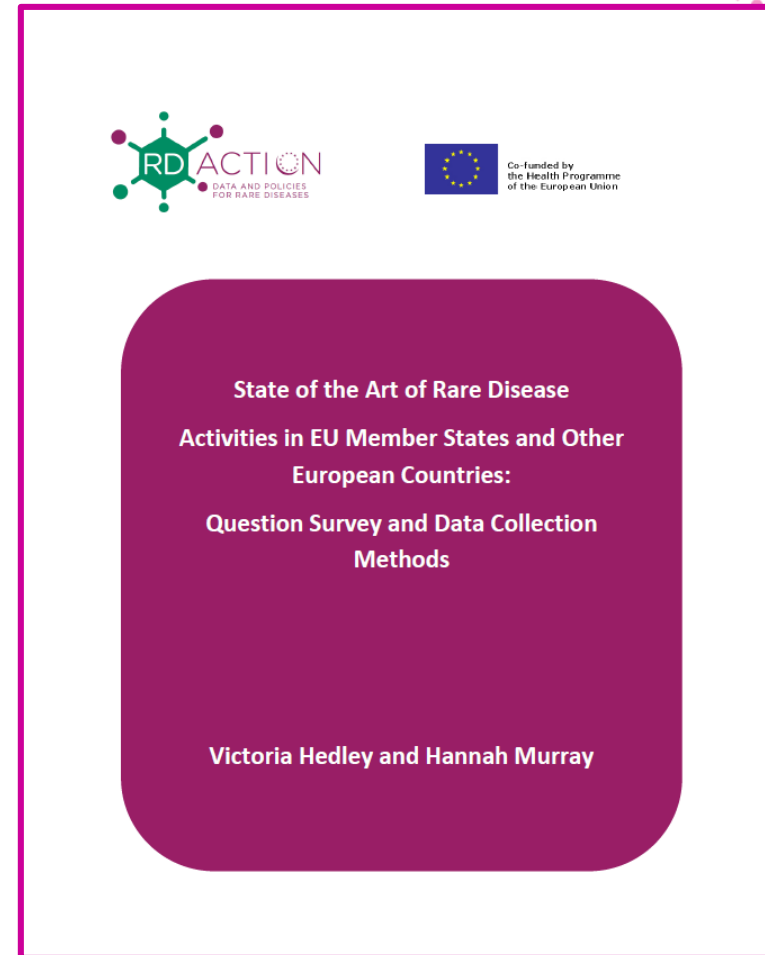
RARE
DISEASES
INTERNATIONAL

CONTENTS

1. Introduction
2. Question Mapping
 - A. National Plans and Strategies for Rare Disease
 - B. Rare Disease Registration
 - C. Definition of a Rare Disease
 - D. Neonatal Screening
 - E. Genetic Testing
 - F. Centres of Expertise
 - G. European Reference Networks (ERNs)
 - H. National Alliances of Patient Organisations and Patient Representations
 - I. National Helplines for Rare Disease
 - J. Activities of Orphanet/Alternative RD-specific information systems
 - K. Official Information Centres for Rare Diseases
 - L. Clinical Practice Guidelines (CPGs)
 - M. Training and Education
 - N. Rare Disease Events
 - O. Existence of Rare Disease research programmes/projects in your country
 - P. Participation in E-Rare and International Research Initiatives.
 - Q. Orphan Medicinal Products (OMPs)
 - R. Social Services and Social Integration
 - S. Other

COUNTRY DATA - METHODOLOGY

- To be more inclusive, set-up Data-Contributing Committees
 - MS representative of EU Commission Expert Group on RD
 - Orphanet Rep
 - National Alliance Patient Rep
- Link sent to DCC to fill out online survey (twice a year)
- Multi-Person Survey electronic tool
- Ultimate responsibility of MS rep in EU CEGRD
- Question bank based on EU CERD Recommendations on core indicators of NPRD + UNEW RD-ACTION team + EU DG Health, DG Research, DG Employment & Social Affairs



RARE
DISEASES
INTERNATIONAL

WHAT IT LOOKS LIKE



CONTACT | INTRANET

Home About us Directory Workpackages News Leaflet and documents



CHANGE COUNTRY

AUSTRIA

ORPHANET-AUSTRIA

NATIONAL PLAN

DOWNLOAD state of art :

2012

2013

2014

End of 2016

Austria

Visual Recap

Contributor's names

Summary of Rare Disease Activities in Austria*

[\(Please see below for link to most recent FULL Country Report\)](#)

Austria formally adopted the European Commission definition of a rare disease when it approved a National Plan for Rare Diseases in February 2015. The NP was developed by the National Coordination Centre for RD (CCRD) which was established by the MoH in April of 2011. The CCRD works closely with two committees established to support its operations: the Expert Committee on RD (which has a very strong multistakeholder composition), and the Strategic Platform



RARE DISEASES INTERNATIONAL

WHAT IT LOOKS LIKE



CONTACT | INTRANET

Home About us Directory Workpackages News Leaflet and documents



contents; the findings and conclusions do not necessarily represent the views of the European Commission or national health authorities in Europe. Therefore, no statement in this report should be construed as an official position of the European Commission or a national health authority

Information supplied by the Austrian SOA Data Contributing Committee:

- Competent National Authority – Till Voigtländer
- Orphanet National Representative – Ursula Unterberger
- National Patient Alliance Representative – Ulrike Holzer

DISCLAIMER

This website is part of the project / joint action '677024 / RD-ACTION' which has received funding from the European Union's Health Programme (2014-2020).
The content of website represents the views of the RD-ACTION consortium only and is its sole responsibility; it can not be considered to reflect the views of the European Commission and/or the Consumers, Health, Agriculture and Food Executive Agency or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains.

MENU

Home
About us
Directory
Workpackages
News
Leaflet and documents

SOCIAL NETWORK

SEARCH



Design & Création : Agence la naissance



RARE
DISEASES
INTERNATIONAL

ADDED VALUE OF THE SOA NATIONAL INFORMATION

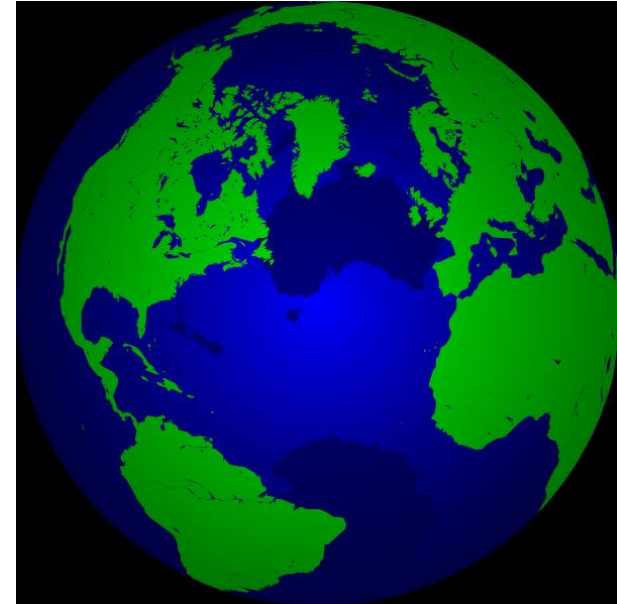
- Provides clear, accurate perspective of the realities (strengths and challenges) in all EU MS
- Ability to make comparisons between countries over time
- Enables statistical analysis on trends (e.g. around national registries, coding, evaluation of NP/NS etc,)
- Cross-country analysis and short SoA 'snapshots' by topic (e.g. Status Quo of NPRD in Europe; Newborn Screening; Centres of Expertise; Registries)
- Helps assess the extent to which Recommendations are being implemented on the ground (and propose new activities)
- Greater transparency and closer relationships between stakeholder groups via the DCCs



BEYOND EU 28

PROPOSAL:

- RDI might take an active role to develop the SoA in other countries around the world beyond EU.
- The approach would be step wise and very pragmatic.
We would start it in few countries where there is a strong willingness and someone ready to do the work, using similar methodology as in EU.
- Newcastle University could provide the survey tool and establish timelines
- RDI could compile the report and summary
- Still ascertaining exactly where to show the information – RD-ACTION site ideally
- Will involve setting-up a DCC in participating countries



RARE
DISEASES
INTERNATIONAL

HOW COULD WE EXTEND IT BEYOND EU?

► Need a DCC:

- National Alliance of RD Patients
- Orphanet / RD database
- National Competent Authority
- MoH – RD focal point/ contact
- Research agency – IRDiRC
- National Pharmaceutical Association
- Other?



THINK ABOUT THESE QUESTIONS

- ▶ How would I use a State of the Art of Rare Diseases to advocate in my country?
- ▶ How could I collect and verify this type of information in my country?

