Resource on the State of the Art of Rare Disease Activities in Europe

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This presentation is part of the project / joint action ‘677024 / RD-ACTION’ which has received funding from the European Union’s Health Programme (2014-2020).

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Background to SoA Report

• ‘State of the art of RD activities in Europe’ report
  – Conceived in 2011
  – Originally funded under EUCERD Joint Action
  – ‘Go to’ source of information on RD activities
  – 3 editions (2012, 2013, 2014), each one 5 volumes (around 500 pages)
    • Plus individual country reports
  – Around 15’000 downloads (all volumes combined) annually
  – Contributions from over 100 stakeholders across Europe
RD-ACTION

RD-ACTION Key Stats

• 1\textsuperscript{st} June 2015 - 31\textsuperscript{st} May 2018
• 34 beneficiaries (APs)
• 30 collaborating partners
• 40 countries incl. Argentina and Australia
• Total Budget: €8,344,079.80
State of the Art Resource: RD-ACTION

• INSERM led the production of this excellent annual Report for 3 years
• Good info but v. lengthy reports, resource intensive
• Now more of a web-based resource, composed of
  – Two overview reports across the three years 2015-18
  – One dedicated webpage per EU MS*
• Differences:
  – More multi-stakeholder contributions
  – Collection of data via a survey
  – Providing a summary but also providing links to the very valuable archived data
  – refresh the information more frequently
Building the Questionnaire (1)

• Wanted something that would collect comprehensive data but also captured key facts for all – so mixture of ‘do you have’ (Y/N questions) and ‘what do you have’?

• RD are complex, and a strong national programme will, in time, ideally have measures in many areas to ensure appropriate diagnosis, treatment and care

• So, many topics maintained with few mandatory Qs for each:
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
• Goal to streamline data collection and optimise use of the data collected

EUCERD had adopted Recommendations on Core Indicators for RD NP/NS – Largely process indicators but MS had committed

• Goal to somehow assess the impact of the various sets of EUCERD and CEGRD Recommendations:
Country Data - Methodology

• To be more inclusive, WP6 set-up Data-Contributing Committees (MS representative, Orphanet Rep, National Alliance Rep) – summer 2016
• Link sent to Data submitted via online survey
• 1st set of data requested for end Sept
• ‘Teething’ troubles!
  – Confusion over concept
  – Not reading the survey instructions
  – Resources on side of the MS reps
  – Need to engage different actors
SOA Survey Technical Guidance

Thank you for completing this survey – your contributions are greatly appreciated. The survey tool is very simple to use and intuitive: there are only a couple of things you need to remember😊. This guidance aims to escort you through the process of answering the survey questions and to steer you through any problems which may arise.

If you work your way through the guidance notes and still cannot resolve your query please contact Hannah Murray (Hannah.skingsley@newcastle.ac.uk).

1. The CEGRD representative will receive an email from us (this will come from Hannah.skingsley@newcastle.ac.uk) which will provide you with a link to the survey. This survey link will be specific to your country.

2. The CEGRD representative (in whose name the link will have been generated) will need to enter the survey as soon as possible by clicking on the link OR copying the link and pasting it into your browser (either way, LIME survey seems to work best with Google Chrome*). You will see the following welcome screen:

![Image of survey welcome screen]

State of the Art of Rare Disease - Activities in EU Member States and Other European Countries

This information is collected as part of the activities of the Joint Action for Rare Diseases, RD-ACTION. The survey is designed to capture important information from all EU Member States (and additional countries) regarding the range of rare disease-related activities implemented or planned. The questions have been compiled based primarily on:

1. The prompt sheets developed by Charlotte Rodwell and utilised under the EU EERD Joint Action and completed by representatives of each Member State once per year.
2. The EUCERD Recommendations on Core Indicators for Rare Disease National Plans/Strategies (containing 1-3 Member State representatives agreed to collect at least annually).
3. Additional/replacement questions devised by the UNEV RD-ACTION team and European Commission experts at DG Sante, DG EMPL and DG RTD to enable an evaluation of the impact of certain policies (e.g., Recommendations).

Ultimate responsibility for completing the survey (twice per year) rests with the Member State representative of the Commission Expert Group on Rare Diseases (or its successor). It is however important that all members of the Data Contributing Committees are able to provide input (please see the dedicated technical guidance on multi-person survey use).

The information, once submitted, will be summarised and synthesised and presented on the new State of the Art webpages which are hosted within the overarching RD-ACTION website (www.rd-action.eu). In completing the survey and submitting the information, the user consents to the use of the data for this purpose alone. Please bear in mind that the information submitted should naturally constitute an accurate summary of rare disease activities in your country. The 'raw' data as provided via the survey will be held and curated by the RD-ACTION team in Newcastle. It will be used solely for population of the State of the Art country-specific pages, the State of the Art overview pages, and the State of the Art overview annual report associated with State of the Art activities. This work is part of project RD-ACTION/07/024, which receives funding from the European Union's Health Programme 2014-2020.

If you have any issues regarding the survey please in the first instance consult the dedicated technical guidance. If the document is unable to answer your question please contact Hannah Murray (Hannah.skingsley@newcastle.ac.uk).
A. National Plans and Strategies for Rare Disease

**Question:**
Has your country adopted a National Plan/Strategy for rare disease?

**Answer:** (Tick Box)
Yes

**Answer:** (Tick Box)
No

**Question:**
Does the NP/NS have a time-frame (e.g. 2015-2020)?

**Answer:**
Free Text Comments Box

**Question:**
Please describe the formal approval process for this NP/NS (e.g. is the NP/NS enshrined in any laws or decrees)?

**Answer:**
Free Text Comments Box

**Question:**
Financial Support for the NP/NS
Is there a policy-decision to ensure long-term funding/sustainability of the measures in the NP/NS?

**Answer:** (Tick Box)
Yes – a dedicated budget exists for the measures in the NP/NS

**Answer:** (Tick Box)
Yes – a policy/decision has been made to ensure long-term sustainability

**Answer:** (Tick Box)
No – there is no associated funding
O. Existence of RD research programmes/projects in your country

**Question:**
Does your country have any specific programmes/projects to fund/facilitate rare disease research?

**Answer:**
- **Yes**
- **No**

**Question:** (tick which best applies):
- a) There is a specific research **programme** for RD
- b) Specific Projects for RD are funded from the **general** research budget

**Answer:**
Free Text Comments Box for further details

**Question:**
Has there been a policy decision to allocate a portion of the national research budget specifically to RD research?

**Answer:**
- **Yes**
- **in development**
- **No**

**Question:**
Please summarise the status quo of rare disease in your country.

**Answer:**
Free Text Comments Box

Please proceed to next section
Added value of the SoA national information

• Provides clear, accurate perspective of the realities (strengths and challenges) in all EU MS
• Greater transparency and closer relationships between stakeholder groups via the DCCs
• Enables statistical analysis on trends (e.g. around national registries, coding, evaluation of NP/NS etc.)
  – Topic-specific short reports to be created this summer
• Should also allow an assessment of the extent to which Recommendations are being implemented on the ground (and propose new activities)
Where are we now?

• Began with the EU 28 MS
• No data submitted from France, Luxembourg, Estonia for 2016
• A few provided very incomplete data (Greece, Poland, Ireland), but good data for 20
• Each has a report, which is being sent to DCC for quick approval
• Then each page will go ‘live’...
Private: RARE DISEASE POLICIES IN EUROPE

Description of the report
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Country reports/block
Select a country to view this report:

key figure & diagram/block

reports & links block
Timelines and Next Steps

• If all reports are approved, each MS submitting data will have page with the summary and stats & link to longer report by end of June

• June and July – cross-country analysis and short SoA ‘snapshots’ by topic

• July – end Sept: the DCCs will be invited to review and update via the portal.

• Will aim to get a TC with each DCC this time

• Will chase-up to obtain more details on statements
Beyond the EU 28?

• As a separate exercise, the Overview report (submitted but formally publicly available later in June) includes report on policy frameworks outside of Europe.
  – Inclusion and data based largely on OrphaNews articles and developments.
  – Not possible to verify and run past the countries (who??)– but if you see errors, let me know

• However... Began discussions with EURORDIS and now RDI on expanding the country data-collection somehow
Beyond EU 28

PROPOSAL:

• EURORDIS suggestion that 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 the exact same methodology as in EU.

• Will involve setting-up a DCC in 4 or 5 countries (Canada, Russia, China, Australia)

• We can provide the survey ‘instance’ and establish timelines

• RDI could compile the report and summary

• Still ascertaining exactly where to show the information – RD-ACTION site ideally
Key Messages

• SoA Resource (several things) remains an invaluable resource for RD field
• Full suite of outputs will be available by the end of the Summer and updates encouraged dynamically henceforth
• We would very much like to expand the scope, and open up the tool to more countries – because RD are a global issue!!
• However – some challenges... no CEGRD, hence constituting those DCCs is difficult
• Resources! To deliver the European material, you need a person working FT (combo of policy experience & administrative support)