RD-ACTION proposal

Promoting Implementation of Recommendations on Policy, Information and Data

For Rare Diseases

Ana Rath and RD-ACTION participants

Rare diseases, an European Priority

- November 2008
  - **Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare diseases: Europe's challenges COM (2008)**
- June 2009
  - **Council Recommendation on an action in the field of rare diseases (2009/C 151/02)**

Rare diseases, an European Priority

- A continuous work on policies for RD
  - Rare Diseases Task Force
  - European Union Committee of Experts on Rare Diseases (EUCERD)
  - Commission Expert Group on Rare Diseases (CEGRD)
- A database for RD that becames European and global
  - Orphanet (EU co-funded since 2000)
- A dedicated nomenclature for RD
  - ORPHA nomenclature
  - Preparing ICD11

Objectives

- To continue **implementation of the policy priorities** identified in Council Recommendation 2009/C151/02 and the Commission Communication (COM 2008 679) on RD, with a view to ensuring the sustainability of the recommended priority actions, and to support the work of the Commission Expert Group on Rare Diseases by gathering expertise and producing data necessary to its action.

- Contribute to solutions to ensure an appropriate **codification** of rare diseases in health information systems

- Support the further development and sustainability of the Orphanet **database** on rare diseases which is run by a large consortium of European partners and is the biggest global repository of information about rare diseases.

RD-ACTION coverage

- Partners in EU were officially nominated by their Ministries of Health

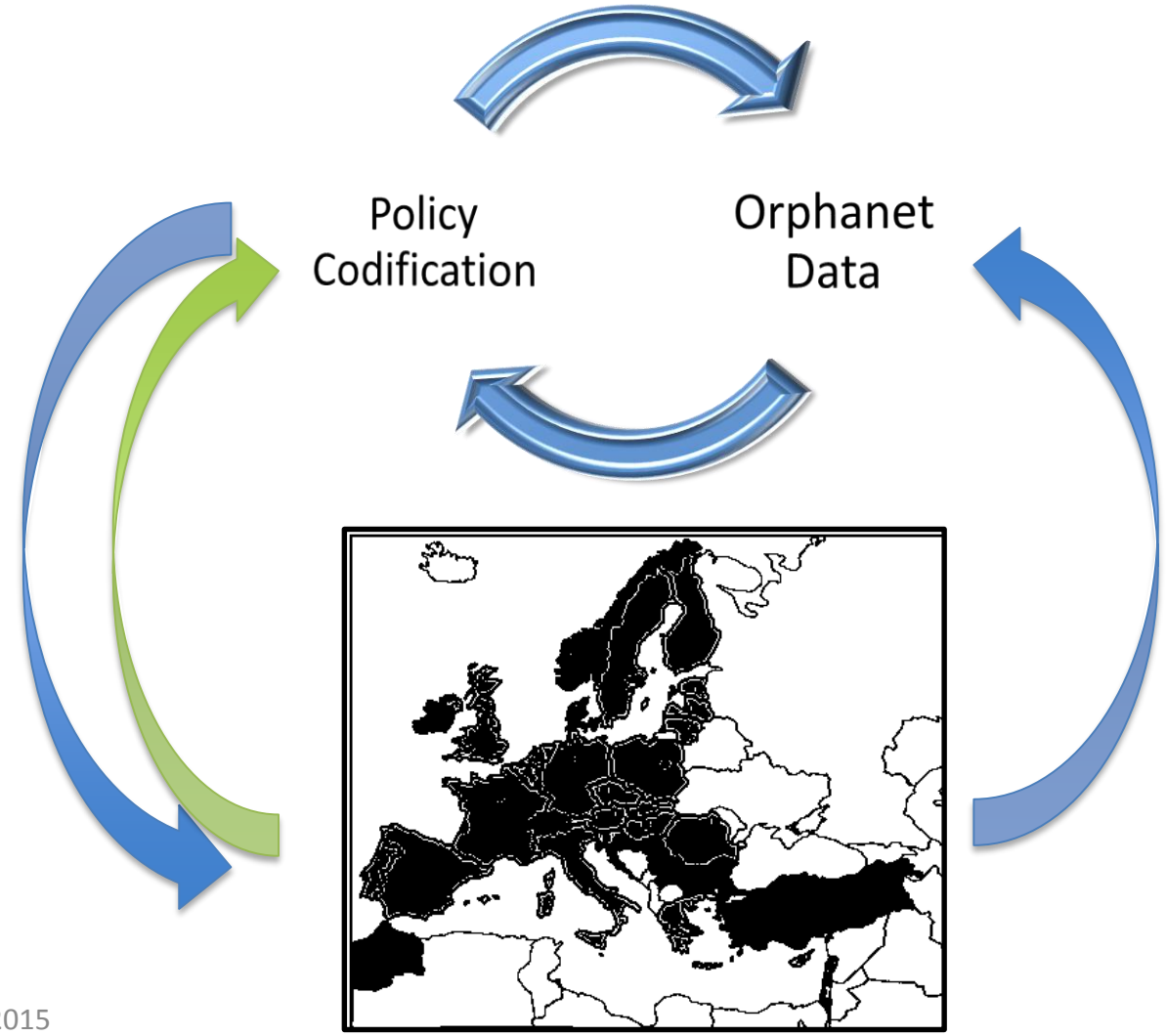
- 53 partners (beneficiary and collaborating)
  - Complete European coverage (excepted from Danemark)
- 11 collaborating partners outside EU

- Italy is represented by 3 institutions:
  - Bambino Gesu Hospital (Bruno Dallapiccola; Orphanet Italy)
  - Veneto region Italian Interregional Board for Rare Diseases (Paola Facchin, Codification)
  - Istituto Superiore de Sanità (Domenica Taruscio, Dissemination)

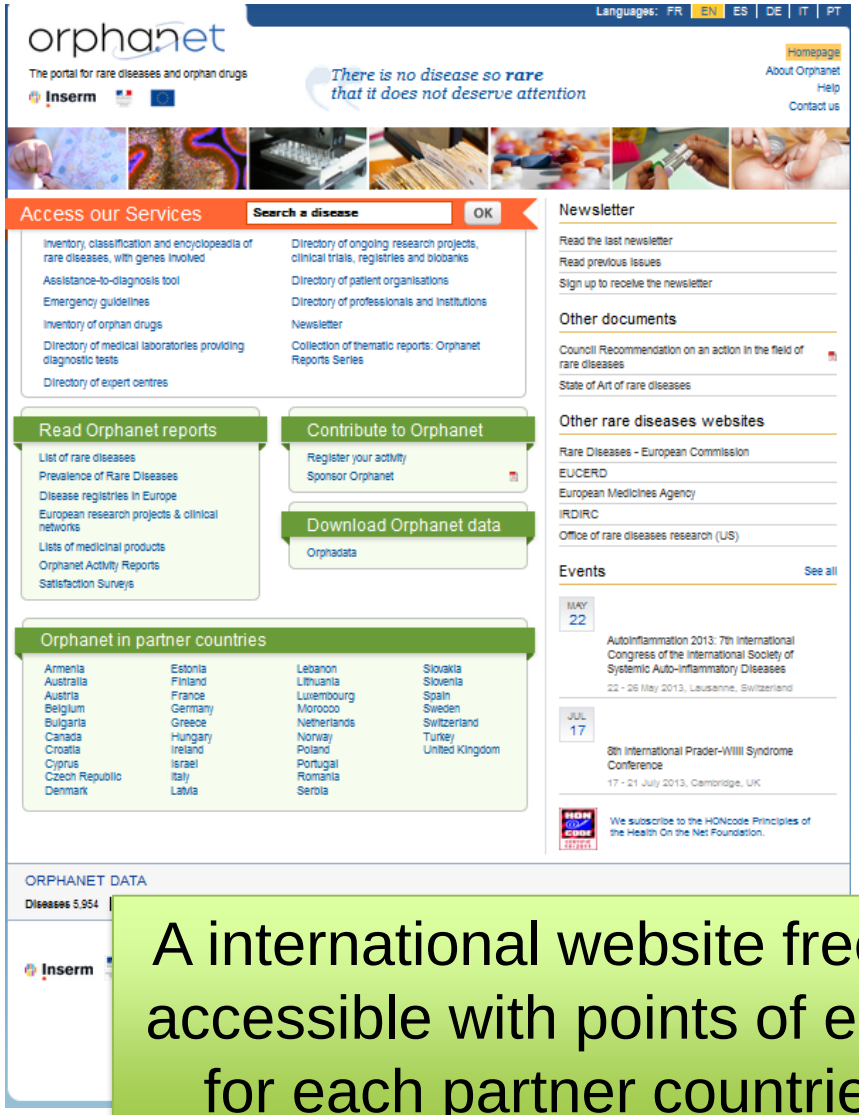
RD-ACTION structure

- Transversal WPs
  - WP1: Coordination (INSERM)
  - WP2: Dissemination (EURORDIS, co-leader ISS)
  - WP3: Evaluation (Medical University of Vienna)
- Core WPs
  - WP4: Orphanet database (INSERM)
  - WP5: Codification (DIMDI)
  - WP6: Policy priorities/Support to CEGRD (UNEW)

Virtuous cycle functionning

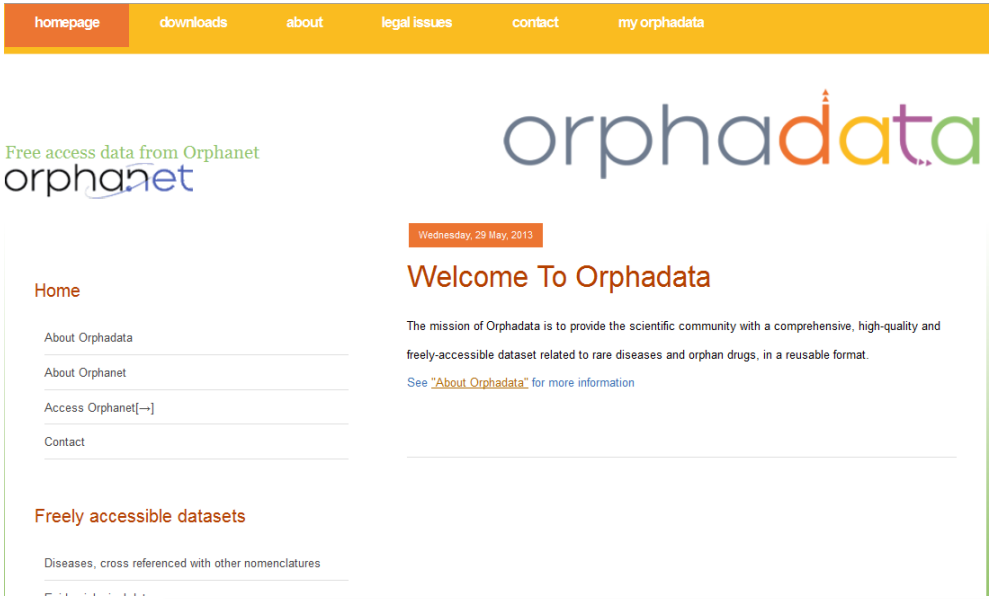


WP 4 - Orphanet



A international website freely-accessible with points of entry for each partner countries

Providing information and data



A download platform of sets of data for re-use

WP4: Orphanet as a EU database

- Maintain and expand the **nomenclature** with cross-references (cross-talk with WP5)
- Expand and update the **encyclopaedia**
- Produce **scientific annotations** for RD (genes, epidemiology)
- Expand and update the directory of **expert resources in participating countries**
- Produce **compiled data** (Orphanet Report Series; cross-talk with WP6)

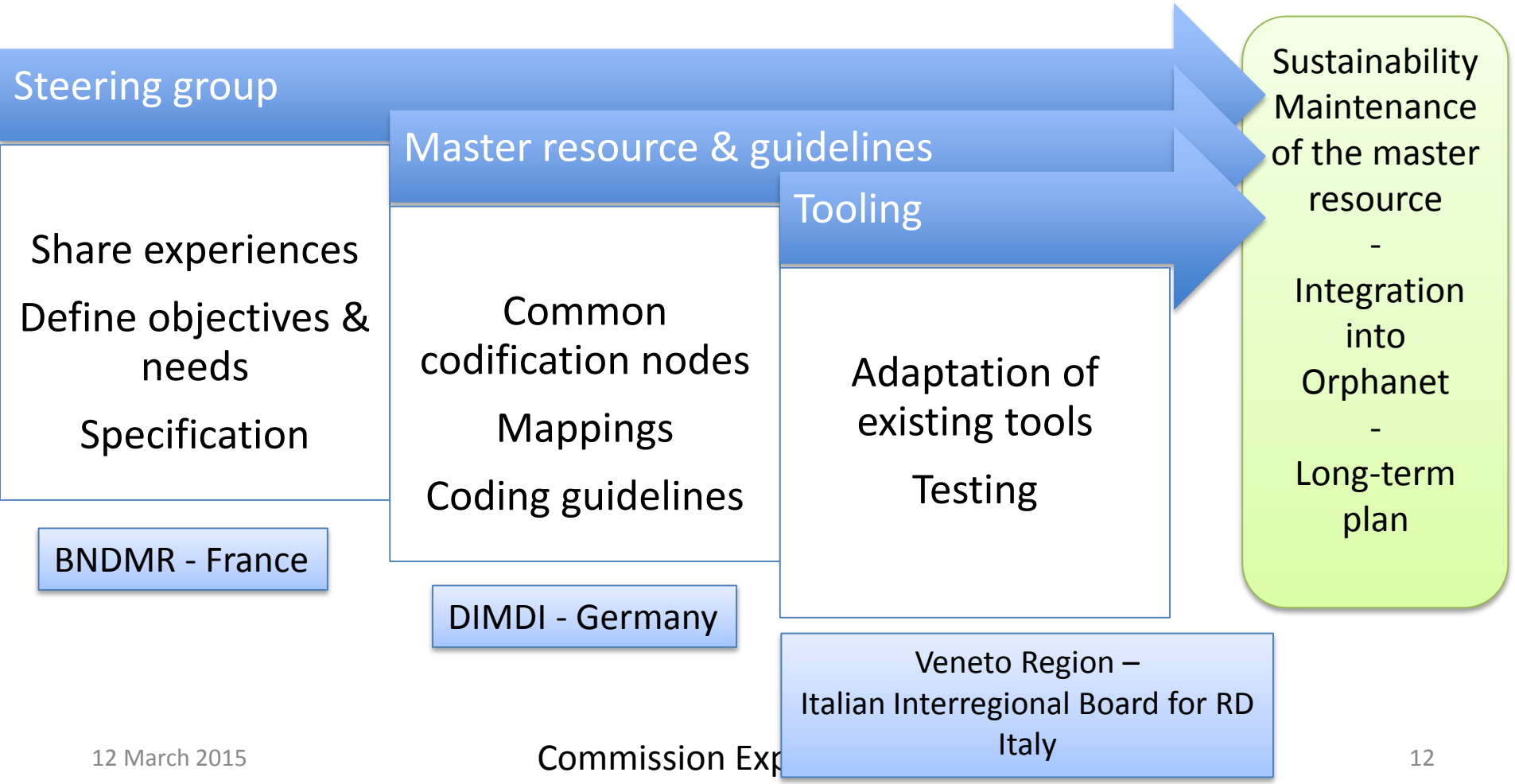
Orphanet as a EU database

- Evolve towards an more open and distributed model
- Improve transparency and traceability of data
  - Web-based knowledge management service to organise the edition and expert-curation

WP3: Focus on Orphanet evaluation & sustainability

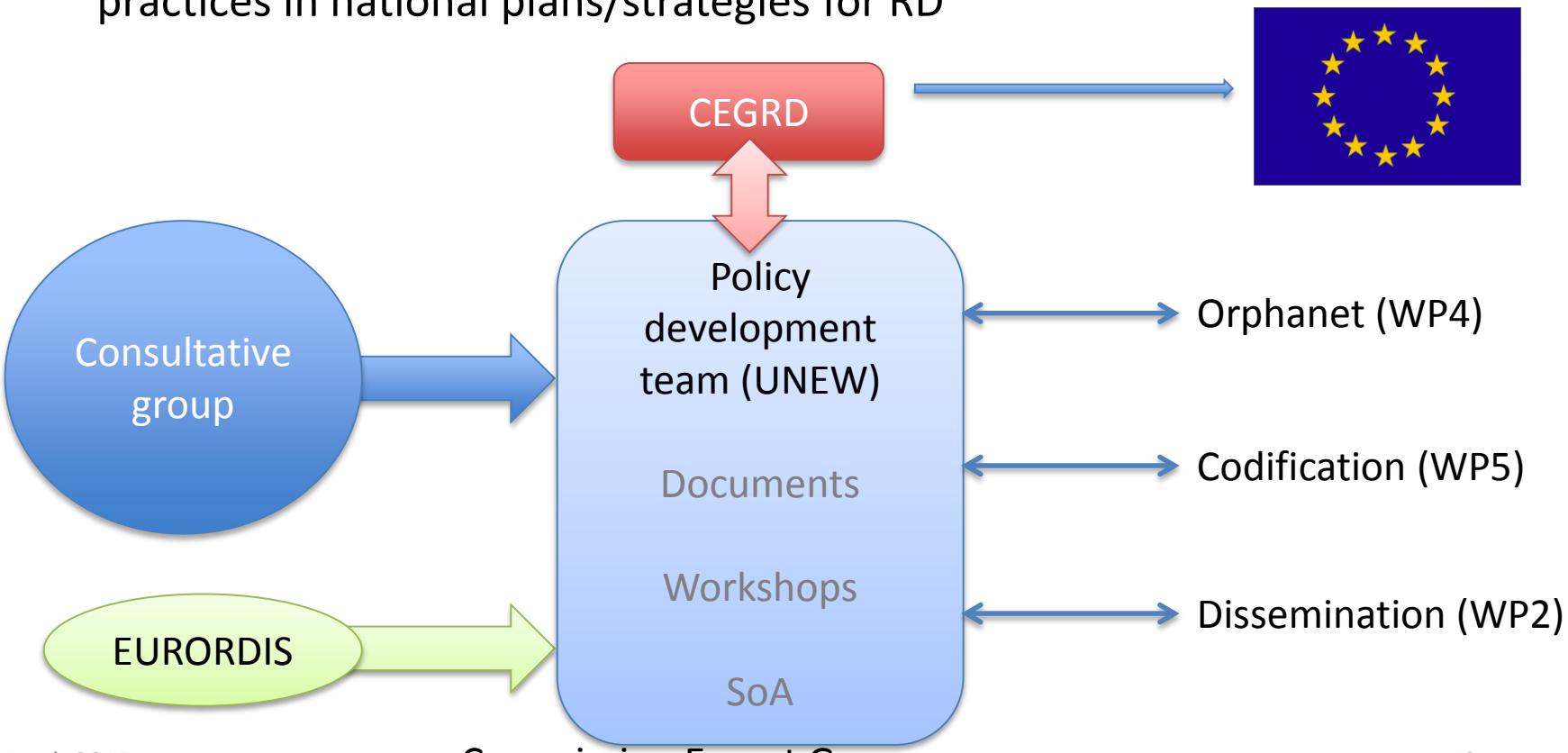
- Organise the evaluation of the Orphanet offer compared to MS needs
  - Users' satisfaction survey
  - Pro-active survey (all stakeholders)
- Set up a sustainability plan
  - Modular representation of Orphanet in view to allow MS to prioritise their needs and support
  - Legal instrument
  - Funding scenarios

WP5: Codification of RD



WP6: Policy priorities

- Objectives:
  - Support the CEGRD work
  - Support the implementation of EUCERD/CEGRD Guidance and Recommendations in MS, and monitor ongoing progress and best-practices in national plans/strategies for RD



Policy topics

- ERNs 2013
- Centres of Expertise 2011
- Registries and Data Collection (including quality issues) 2013
- Integrating rare diseases into social policies
- Genetic testing/screening 2013 and NGS
- Comprehensive information systems
- Generation and sharing of best practice and guidelines on diagnostics and care
- Pricing and access to therapies (HTA)
- e-health
- Prevention of congenital anomalies
- Evaluation of the socio-economic burden of RD.

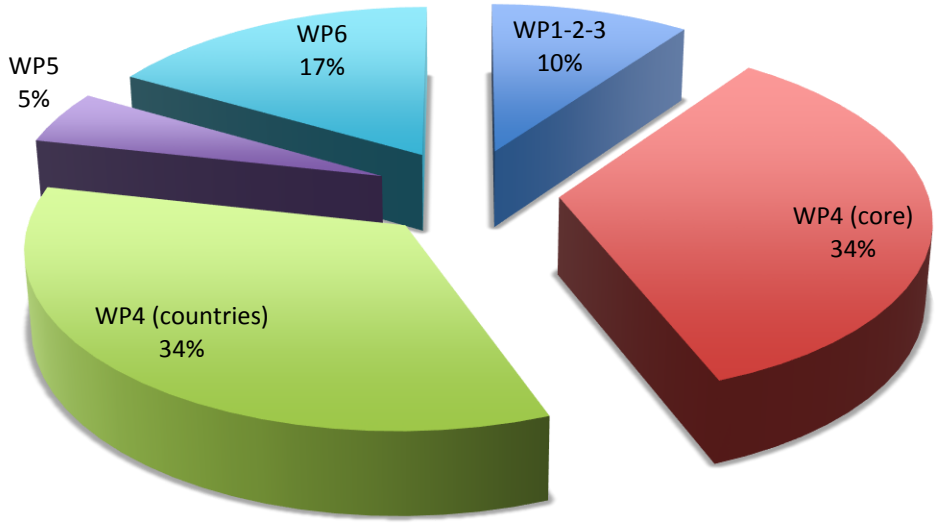
WP6 specific objectives

- Implement a **policy methodology**
- Propose/revise **recommendations**
- Produce **data** to support analysis and decisions (cross-talk with WP4; ORS)
- Develop an electronic **State of the Art** of RD activities in Europe resource
  - To present the SoA
  - To monitor implementation (NP indicators)
  - To allow MS sharing experiences
- Ensure **back-and-forth information flow** between EC and MS (cross-talk with WP2)

WP2: Focus on forth-and-back communication for implementation

- European Conference on Rare Diseases
  - Edinburgh 2016
- Support to national workshops
  - Build on EUROPLAN conferences, methods & experience
  - Focused on European recommendations
  - Accompany their implementation at national level
- Disseminate knowledge on sustainable health systems for rare diseases (ISS)

Budget breakdown



Total EC contribution: 4 380 000 Euros (60%)

Grazie !

Management structure