EURORDIS
Rare Diseases Europe

Registries and Research Infrastructures

Council of European Federations – 29 Oct 2013

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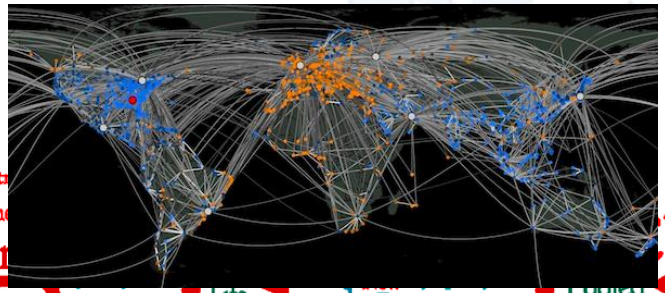
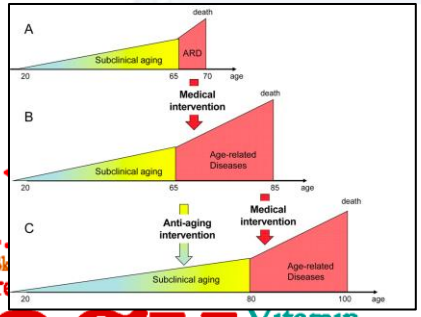
Comprehensive Definition of Registry

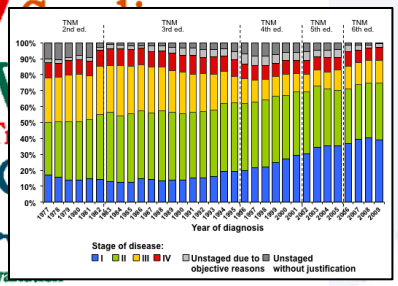
A patient registry is an **organized system** that uses observational study methods to **collect uniform data** (clinical and other) to evaluate specified outcomes for **a population** defined by a particular disease, condition, or exposure, and that serves a predetermined scientific, clinical, or policy purpose(s). The registry database is the file (or files) derived from the registry.

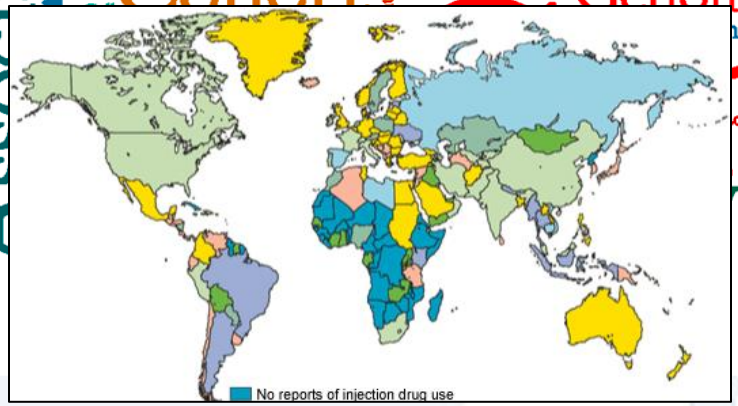
US Agency for Healthcare Research and Quality's (AHRQ) - Registries for Evaluating Patient Outcomes: A User's Guide

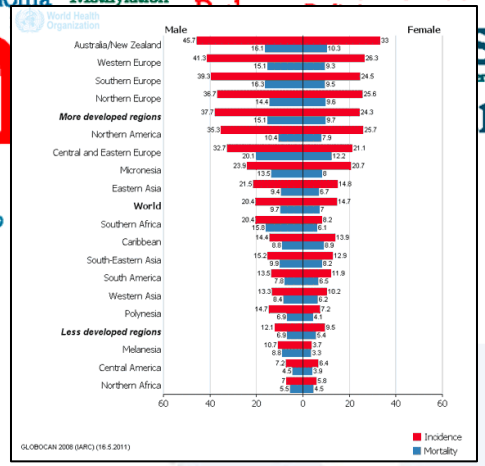
Registries in the Past







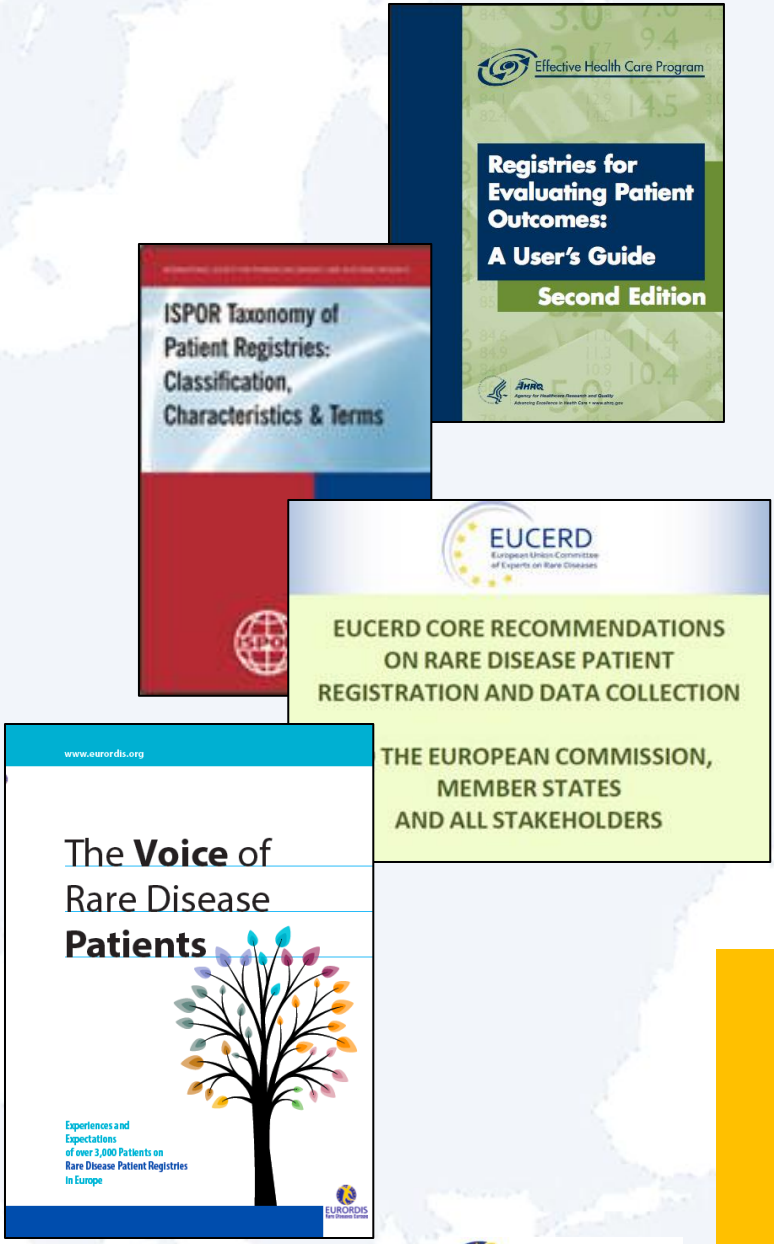




Registries Today

- Perception of registries has changed for scientists, industry, policy makers and patients

- Now key solutions to pulling geographically and structurally dispersed information together



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Value of Registries

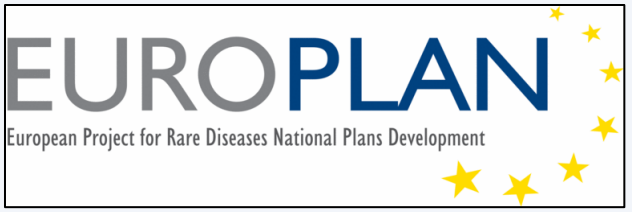
- *Creating a registry of patients is the single most valuable action a rare disease community can take. The registry provides critical disease knowledge which makes that disease easier to study, increasing the probability a treatment can be developed.* - David Meeker, President and CEO of Genzyme, a Sanofi company.

- *Research requires a community of interested-parties to reach the critical mass. Registries, European networks and patient organisations are determinants of research and development for rare diseases.* - Segolene Ayme, Orphanet, RDD 2009 European Workshop

- *Registries are an indispensible infrastructure tool for translating basic and clinical research into improved care and therapeutic solutions.* – Paola Coggi, DG Sanco, Dominique Ristori, JRC, European Commission

- *The importance of rare disease registries cannot be underestimated. Our patient-led register was instrumental in triggering a national enquiry into the cause of death of patients…* - patient organisation representative

European Commission Committment



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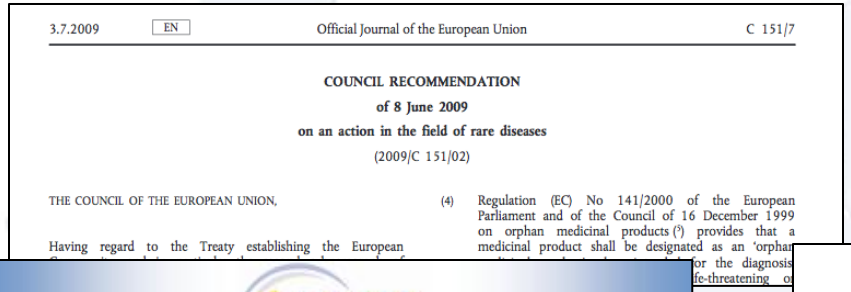
European Recommendations



3.7.2009 EN Official Journal of the European Union C 151/7

COUNCIL RECOMMENDATION

of 8 June 2009

on an action in the field of rare diseases

(2009/C 151/02)

THE COUNCIL OF THE EUROPEAN UNION,

Having regard to the Treaty establishing the European

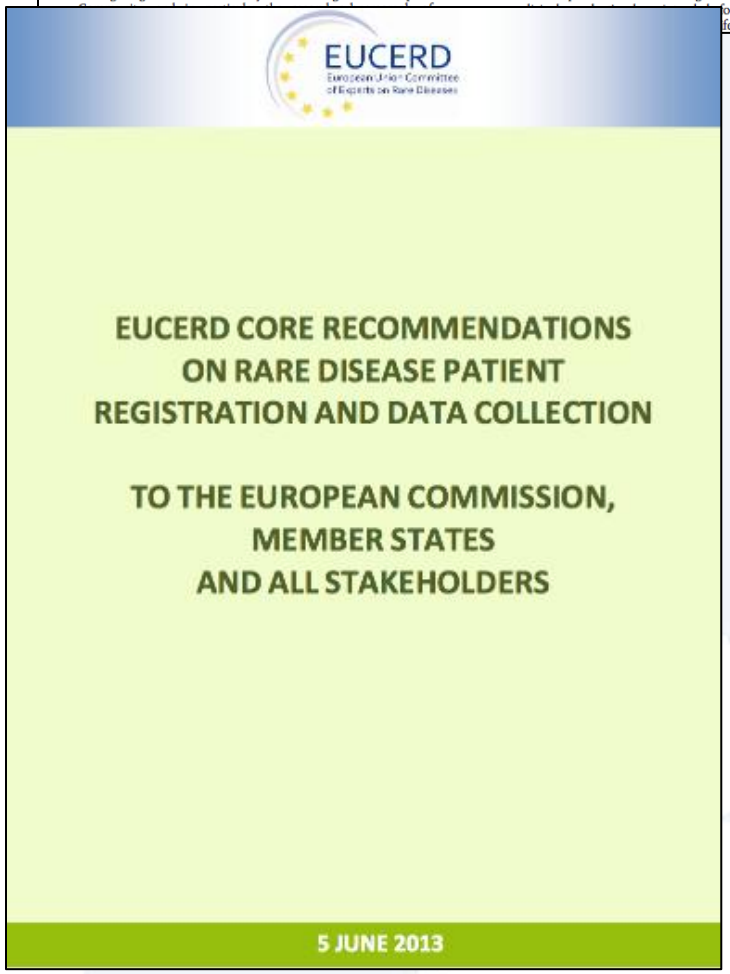
(4) Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products (3) provides that a medicinal product shall be designated as an 'orphan

for the diagnosis

life-threatening o

EUROPLAN

European Project for Rare Diseases National Plans Development



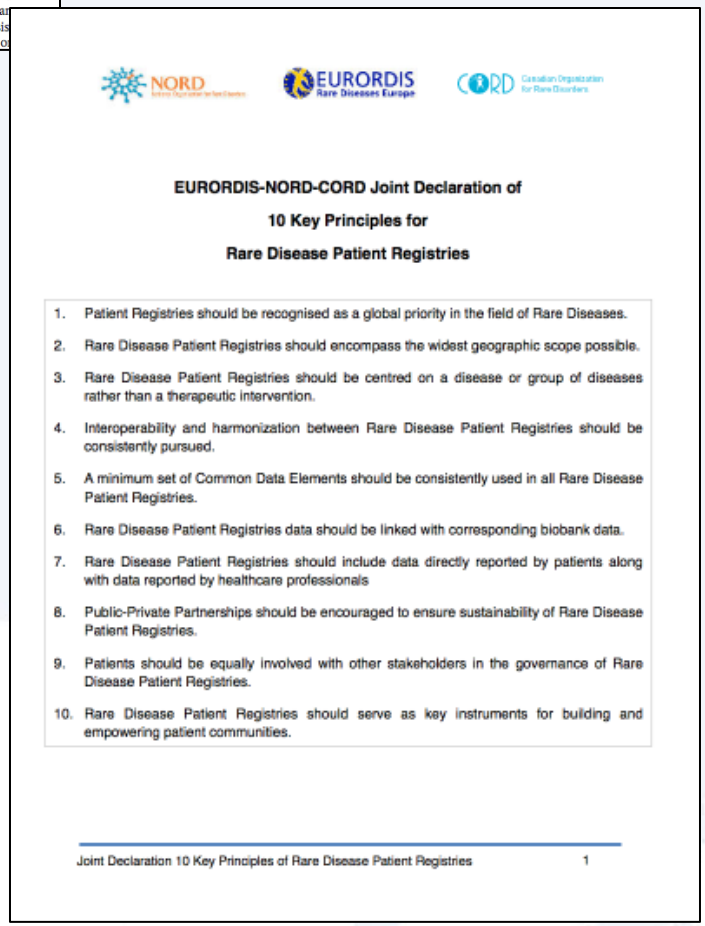
EUCERD

European Union Committee of Experts on Rare Diseases

EUCERD CORE RECOMMENDATIONS ON RARE DISEASE PATIENT REGISTRATION AND DATA COLLECTION

TO THE EUROPEAN COMMISSION, MEMBER STATES AND ALL STAKEHOLDERS

5 JUNE 2013



NORD EURORDIS Rare Diseases Europe CORD Canadian Organization for Rare Disorders

EURORDIS-NORD-CORD Joint Declaration of

10 Key Principles for

Rare Disease Patient Registries

1. Patient Registries should be recognised as a global priority in the field of Rare Diseases.
2. Rare Disease Patient Registries should encompass the widest geographic scope possible.
3. Rare Disease Patient Registries should be centred on a disease or group of diseases rather than a therapeutic intervention.
4. Interoperability and harmonization between Rare Disease Patient Registries should be consistently pursued.
5. A minimum set of Common Data Elements should be consistently used in all Rare Disease Patient Registries.
6. Rare Disease Patient Registries data should be linked with corresponding biobank data.
7. Rare Disease Patient Registries should include data directly reported by patients along with data reported by healthcare professionals
8. Public-Private Partnerships should be encouraged to ensure sustainability of Rare Disease Patient Registries.
9. Patients should be equally involved with other stakeholders in the governance of Rare Disease Patient Registries.
10. Rare Disease Patient Registries should serve as key instruments for building and empowering patient communities.

Joint Declaration 10 Key Principles of Rare Disease Patient Registries 1

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European Platform for Rare Disease Registries

- European Commission has announced strategic objective in creating a European Platform on Rare Diseases Registration at Joint Research Center (JRC) in Ispra, Italy
- Common services and tools for the existing (and future) rare disease registries in Europe.
- 2M EUR over 3 years

European Commission

JOINT RESEARCH CENTRE

Institute for Health and Consumer Protection (IHCP)

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European Platform for Rare Disease Registries

- In the framework of the EPIRARE project
- EURORDIS has proposed policy scenarios on the aims, scope, governance structure and long-term sustainability
  - Aims – harmonise rare diseaes patient registries in Europe
  - Scope – all RD all EU
  - Governance – sound, with patient representation
  - Sustainability – long term… beyond initial funding

Half a Score

An EPIRARE project production.
Written and directed by Gianni Del Corral

RD Connect

The genomic revolution will offer additional opportunities for improving health for patients with RD through advancing research and integrating data from –omics research with phenotypic information (patient registries) and biomaterials (biobanks).

-Hanns Lochmuller and Kate Bushby, Institute of Human Genetics, Newcastle University

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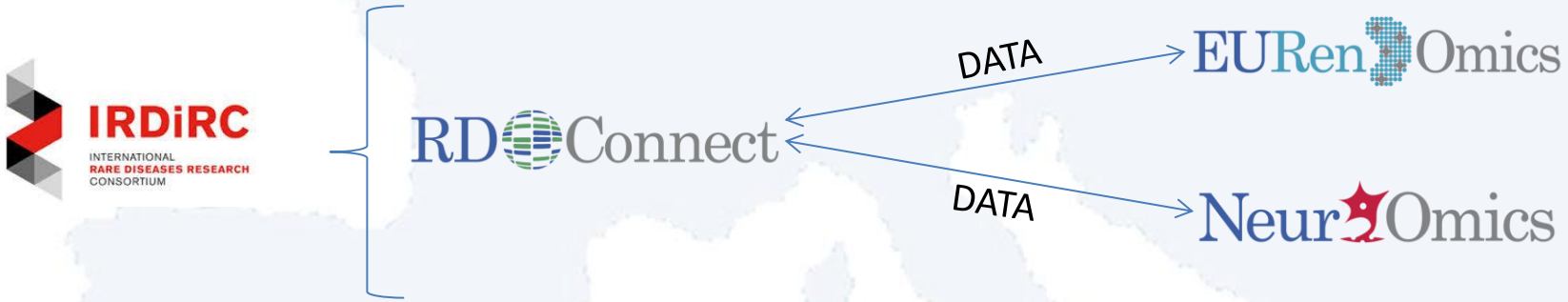
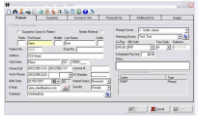
RD-Connect

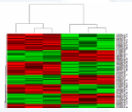
- a unique global infrastructure project that links up databases, registries, biobanks and clinical bioinformatics data used in rare disease research into a central resource for researchers worldwide



Integration of Data

At each point there are data sharing tools and charters

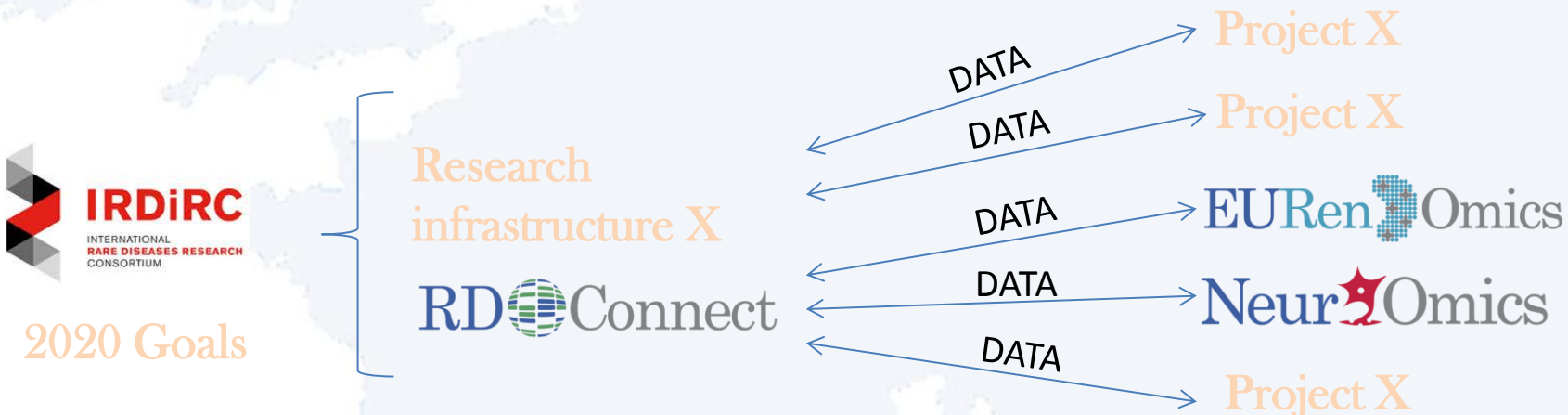




A global unique identifier (ID-RD) will link data from differing sources

With time…

- More projects, more infrastructures… getting us closer to IRDIRC goals



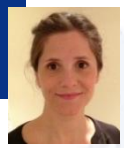
RD Connect Project Objectives

- Harmonisation and development of common standards for databases and patient registries for rare (**WP2**)
- Harmonisation and development of common standards and a common catalogue for rare disease biobanks (**WP3**)
- Development of data mining and knowledge discovery tools for analysis and integration of molecular and clinical data to discover new disease genes, pathways and therapeutic targets (**WP4**)

RD Connect Project Objectives (2)

- Development of an integrated platform to host the processed data from Neuromics, EuRenOmics and future IRDiRC projects (**WP5**)
- Development of best ethical practices by engaging with relevant stakeholders including a proposal for an expedient regulatory framework for linking of medical and personal data related to rare disease on a European and global level (**WP6**)
- Ensuring access to project results and broad and global impact in science, diagnostics and translational research including industrial collaborations (**WP7**)

RD- Connect Who's Who?



Anna Kole, Registry & Biobank Projects Manager, EURORIDS

Yann Le Cam, Chief Executive Officer, EURORDIS

WP2 **WP3** **WP5** **WP6** **WP7**

WP1

Hanns Lochmüller, RD-Connect Coordinator
Professor of Exp. Myology, Newcastle Univ.
Chair of the IRDiRC Interdisciplinary Committee

WP2

Domenica Taruscio, Istituto Superiore di Sanità

WP3

Lucia Monaco, Chief Scientific Officer, Fondazione Telethon

WP4

Christophe Béroud, Head of bioinformatics team at the UMR_S 910 research unit, Aix-Marseille Univ. Medical School

WP5

Ivo Gut, Director of Centro Nacional de Análisis Genómico

WP6

Mats Hanson, Professor of Biomedical Ethics, Uppsala University

WP7

Kate Bushby, Professor of Neuromuscular Genetics, Newcastle Univ. , Co-chair of EUCERD

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Patient representation

- PATIENT INVOLVEMENT = IMPROVED RESEARCH PROGRESS
  - Dynamic dialogue between researchers and patients
  - Inter-understanding increases uptake and acceptance of technologies
  - Researchers know developments are relevant and needed

- Promotes high professional standards and ethical integrity
- Patient representatives as actors, ambassadors and governors builds confidence and trust

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EURORDIS – Specific Aims

1. Capacity building on registries, biobanks and -omics
2. Consulting patient perspective
3. Educational materials for project partners

ETHICAL, LEGAL, SOCIAL ISSUES
WP6

4. Identifying sustainability models for long-term maintenance of registries
5. Encouraging increased participation in biobanks

RESEARCH INFRASTRUCTURE BUILDING WP2 and 3

6. Reporting on the impact of project activities
7. Ensure a strong interaction and coordination of the RD-CONNECT network with other initiatives within and beyond Europe
8. Dissemination of the RD-CONNECT project outcomes at the international level

IMPACT AND EVALUATION
WP7

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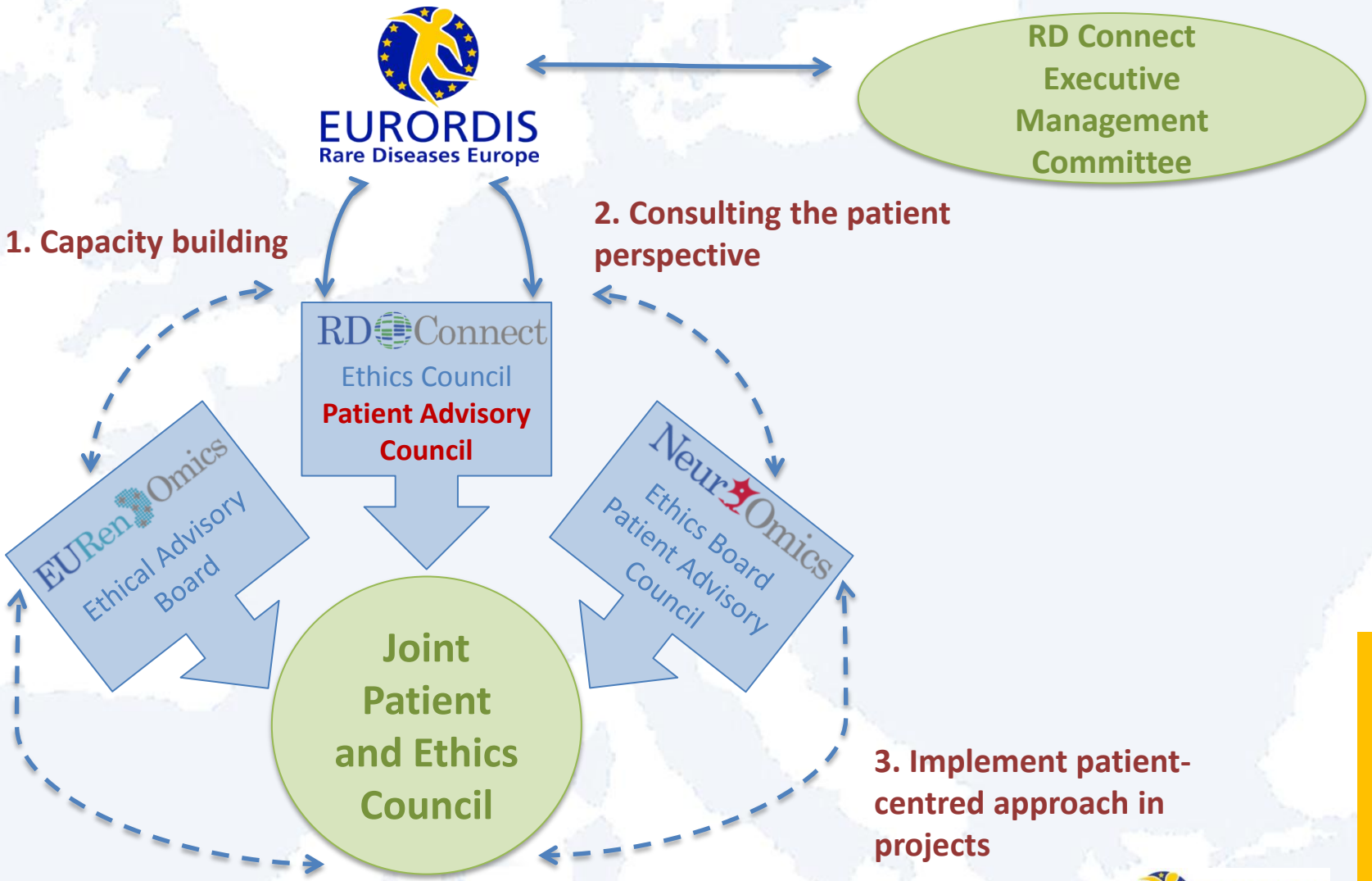
Ethical, Legal and Social Issues



Patients Advisory Council (PAC)

- Voluntary, informal EURORDS Working Group collecting different patient views on issues surrounding registries, biobanking and –omics research
- Advise the RD-Connect Governing Board and the coordinator from the patient perspective
- Initially covering diseases in EuRenOmics and NeurOmics projects, but now extended to others to include diversity in geographic scope and disease characteristics

PAC Who's Who?


Muriel Gevrey, French Charcot Marie Tooth Association


Joseph Irwin, Jennifer Trust for Spinal Muscular Atrophy


Julian Isla, Dravet Syndrome Foundation


Sigurdur Johannesson, Alternating Hemiplegia of Childhood Federation of Europe


Anna Kole, EURORIDS


Dorthe Lykke, European Federation of Hereditary Spastic Paraplegia


Francisco Monfort, European Alliance of patients with Atypical Hemolytic Uremic Syndrome


Kay Parkinson, Alstrom Syndrome UK


Marita Pohlschmidt, Muscular Dystrphy Campaign


Daniel Renault, Federation of European Associations of Patients affected by Renal Genetic Diseases


Françoise Rouault, French Muscular Dystrophy Association - Téléthon


Balthasar Schaap, European Federation of Neurological Associations/EuroAtaxia


Juliette Senecat, EURORDIS


Inge Schwersenz, Spinal Muscular Atrophy Europe


Chris Sotirelis, Spinal Muscular Atrophy Europe

Oliver Timmis, The Alkaptonuria Society


Mariek Van Meel, NephcEurope


Elizabeth Vroom, United Parent Projects Muscular Dystrophy

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Capacity Building

- Workshops alongside project meetings
  - Joint Annual Meetings (Sitges, Heidelberg) – PROJECT AIMS
  - IRDIRC Conference (Dublin) – GLOBAL RARE DISEASE STRATEGY
  - Stakeholder Conference on Regulatory Hurdles (Brussels) – INFORMED CONSENT, DATA PROTECTION AND THE IMPORTANCE OF DATA SHARING
  - European Conference for Rare Diseases (Berlin) – PROJECT OUTCOMES, ENCOURAGING PARTICIPATION
  - EURORDIS Summer School (Barcelona) – FOUNDATON FOR THERAPEUTIC DEVELOPMENT
- EURORDIS Training module
- Advocacy materials
  - Factsheets
  - Online training
- Regular Updates
  - RD Connect website and newsletter
  - EURORDIS newsletter
  - EURORDIS membership events

Consulting Patient Perspective

- Patient Advisory Council (EURORDIS Working Group)
- Literature Review
- Workshops
- Surveys
- Delphi exercise
- Participation in RD Connect, EURenOmics, NeurOmics and Joint Patient and Ethics Council
- Ad-hoc consultations

Get involved

- Opportunity to involve patient voice in a new era of rare disease research where expectations of stakeholders is not fully mature.
- Join EURORDIS Working Group
- Share your experience and expectations and learn from the project outcomes