

IMPACT AND WRAP-UP



Impact session

2

- Feedback from the PAC (Virginie Bros-Facer)
- Short update on IRDiRC
- Short update on ERNs (Vicki Hedley)
- Short summary of sustainability session
- Discussion and Wrap-up



An Update: International Rare Disease Research Consortium (IRDiRC)

Professor Hanns Lochmüller

Chair of Experimental Myology, John Walton Muscular Dystrophy
Research Centre, Newcastle University, Newcastle upon Tyne, UK

IRDiRC Interdisciplinary Science Committee, Chair

Objectives of IRDiRC by 2020

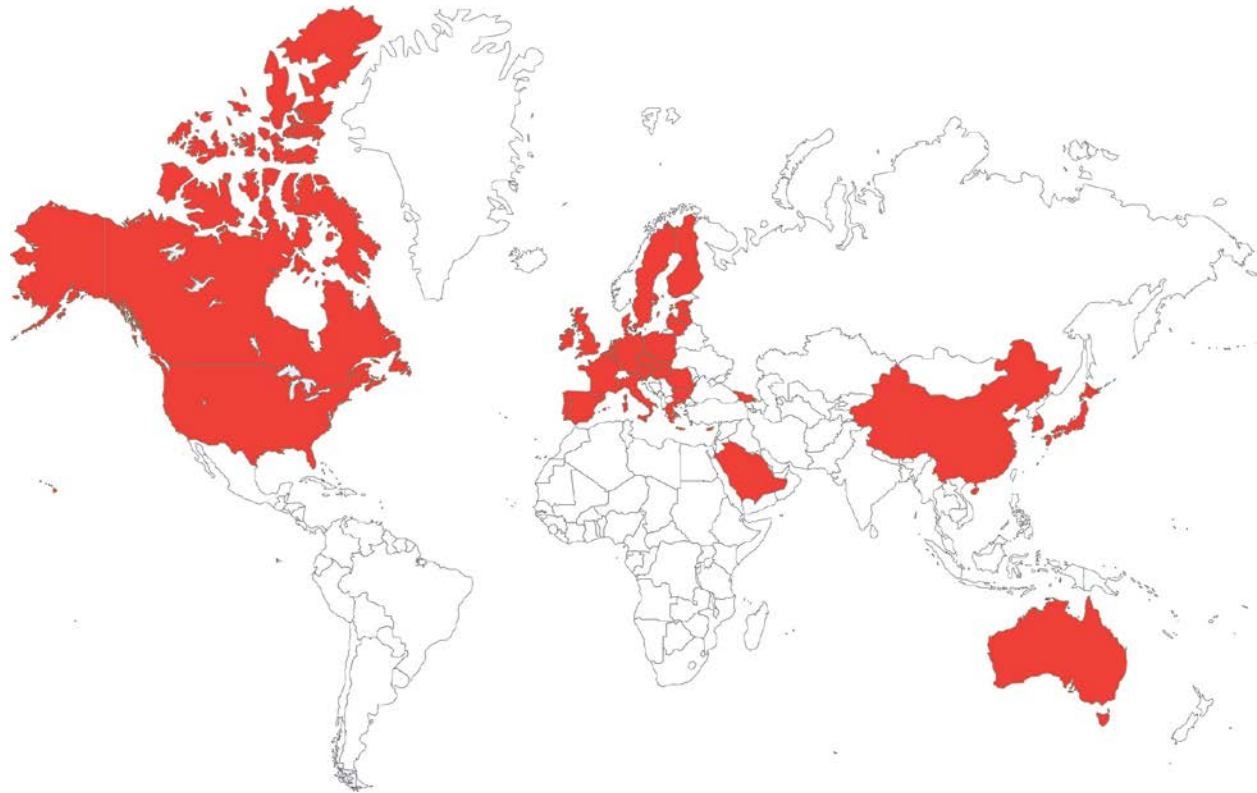
- ▶ 200 new therapies for rare diseases
- ▶ Means to diagnose most rare diseases



IRDiRC's members

► Members from

- Europe
- North America
- Asia
- Australia
- Middle East



IRDiRC

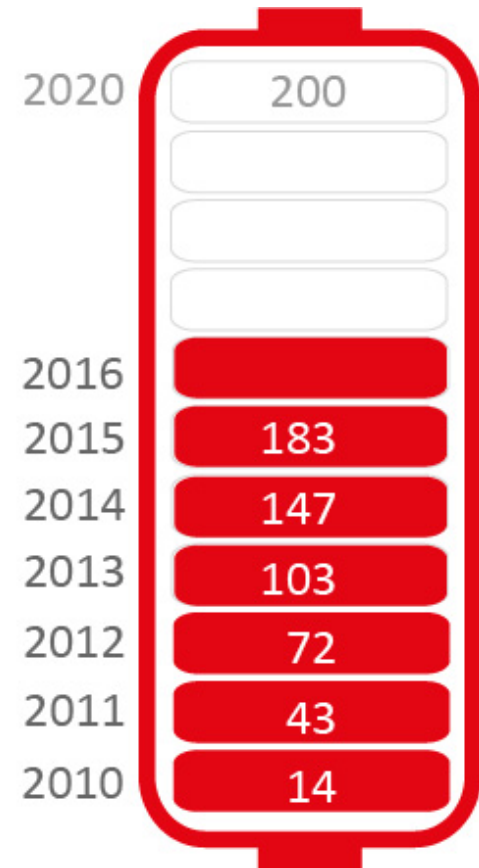
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM



IRDiRC Monitors progress towards goals

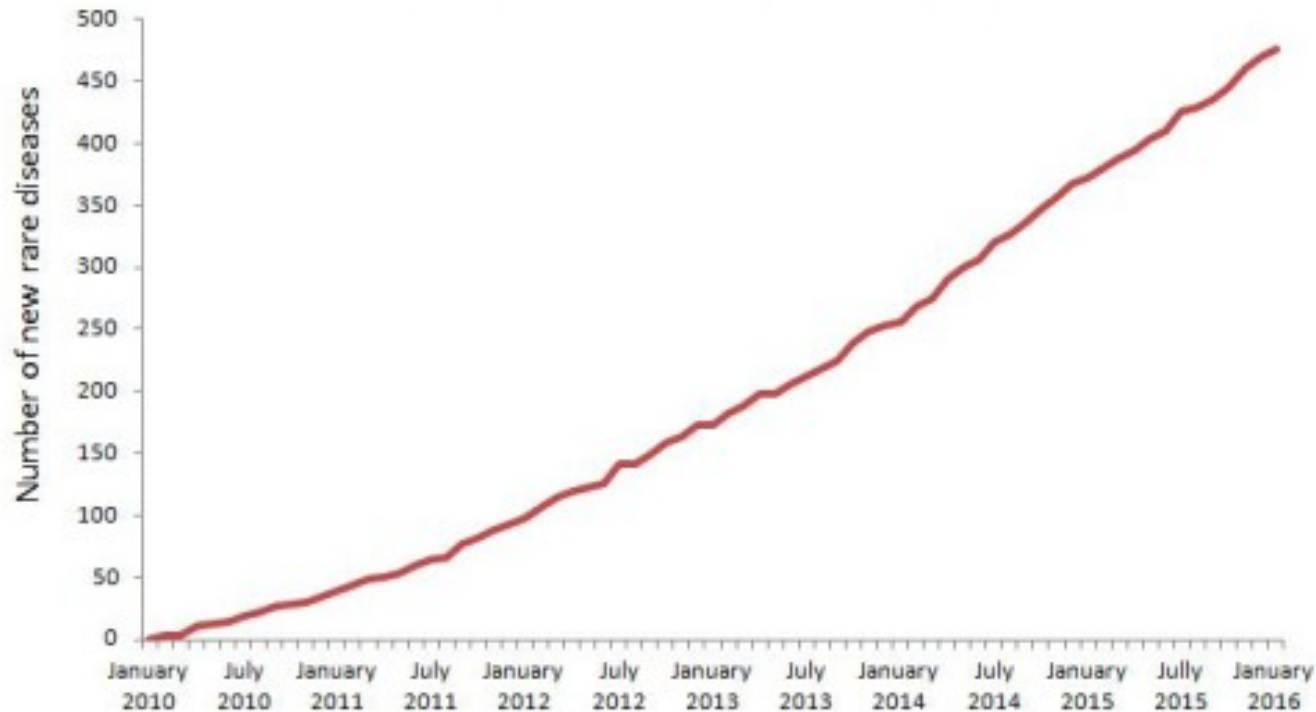
Number of new orphan drugs

- ▶ Monthly updated data are captured into a cumulative and cross-linked table
- ▶ Count in the US (FDA) and EU (EMA)
- ▶ The total is reported into the IRDiRC counter



Number of rare diseases in Europe

Cumulative number of new rare diseases by month since 2010



IRDiRC

INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

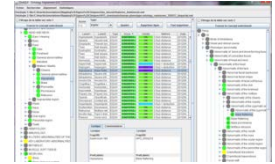
Source: Orphanet Data

IRDiRC Task Forces

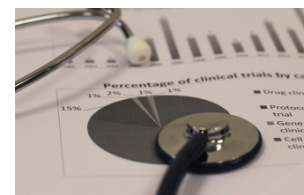
- ▶ To work on selected, actionable topics/research areas and push forward policy change
- ▶ *Ad hoc* committees: nominated experts from different backgrounds, affiliations, geographical areas
- ▶ Collaborate through teleconferences and workshops
 - ↳ Production and dissemination of reports
 - ↳ Implementation of outcomes
 - ↳ Publication in peer-reviewed journals
 - ↳ Presentation at conferences

IRDIRC Task Forces

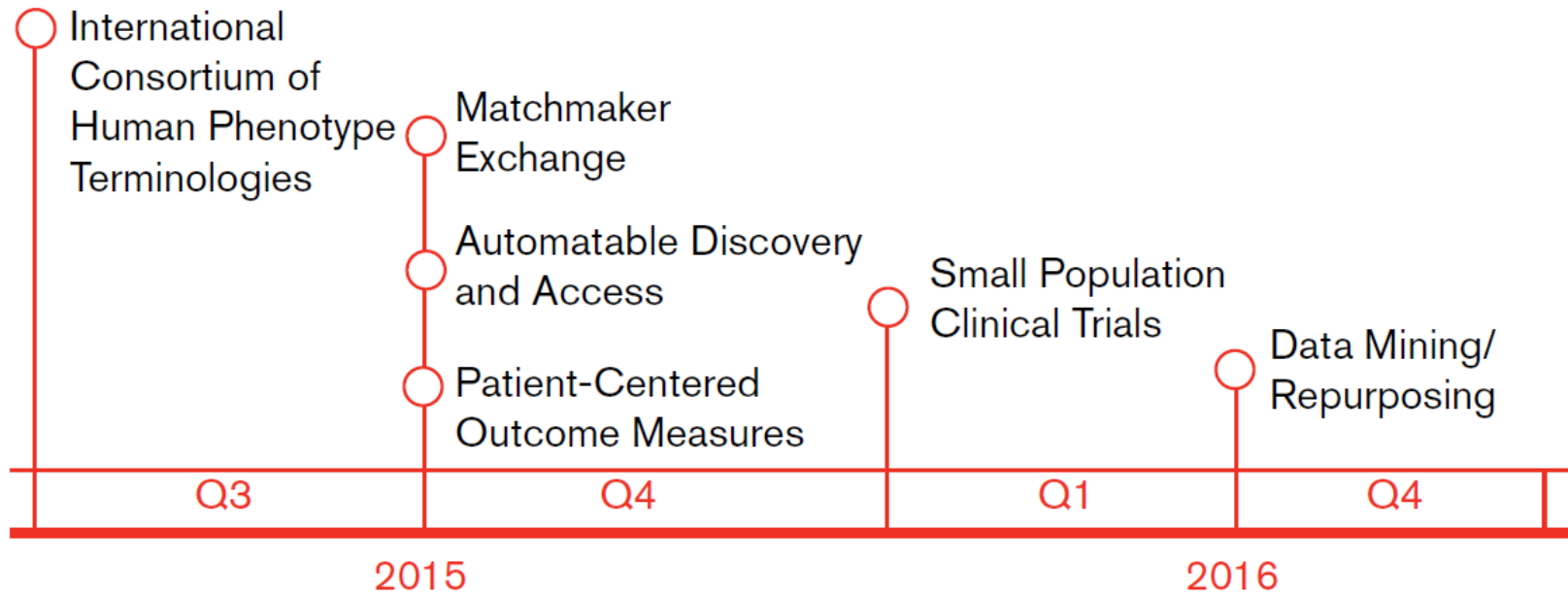
- ▶ International Consortium of Human Phenotype Terminologies
- ▶ Matchmaker Exchange
- ▶ Automatable Access and Discovery
- ▶ Patient Centred Outcome Measures
- ▶ Small Population Clinical Trials
- ▶ Data Mining/ Repurposing
- ▶ Research participant identifier



Matchmaker
Exchange



Timeline for Task Forces



Matchmaker Exchange

- ▶ Provides data sharing tools between clinical geneticists to match unsolved genome/exome sequence cases
- ▶ Ensures optimal collaboration between all projects contributing to the interpretation of variants and of matching phenotypes and variants
- ▶ Joint IRDiRC-GA4GH collaboration



Matchmaker
Exchange

Automatable Discovery and Access

- ▶ Associate clinical data with the scope of consent given
- ▶ Develop standardized and computer-readable data use types in consent forms
- ▶ Aligning a user's permission against permitted data use type
- ▶ Coordinate with the GA4GH and other initiatives (e.g. RD-Connect)



Data Mining and Repurposing

- ▶ Leverage on developments in Computational Linguistics and Graph Theory to build a representation of knowledge which is automatically analyzed to discover hidden relations between any drug and diseases
- ▶ Opportunities for:
 - ↳ Collaborators to exploit data mining tools
 - ↳ Identify new therapeutic targets and repurpose drugs
 - ↳ Increase speed of new drugs available for rare disease patients



Participant Unique Identifier

- ▶ Development of participant unique identifiers for research data sharing across multiple projects and institutions
- ▶ Product: Guidelines on the technical and ethical-legal requirements of patient identifiers in Rare Disease Research; recommendations for the most practical, streamlined and minimalistic approach that maximises uptake whilst complying with relevant legal regulations.
- ▶ Joint IRDiRC-GA4GH collaboration

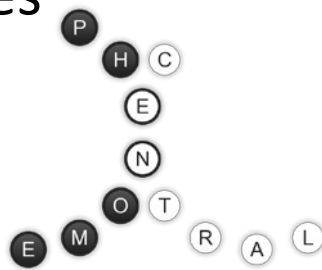


“IRDiRC Recommended”

- ▶ Label highlighting tools, standards, platforms and guidelines which contribute directly to IRDiRC objectives
- ▶ Identification of key resources for research communities to accelerate clinical translation

“IRDiRC Recommended” Resources

- ▶ International Charter of Principles for sharing Bio-Specimens and Data
- ▶ Orphanet
- ▶ PhenomeCentral
- ▶ Orphanet Rare Disease Ontology (ORDO)
- ▶ DECIPHER
- ▶ GA4GH Framework for Responsible Sharing
- ▶ HPO
- ▶ ICHPT
- ▶ TREAT-NMD Patient Registries
- ▶ TREAT-NMD Standard Operating Procedures



orphadata

 **TREAT-NMD**
Neuromuscular Network

orphanet

 **DECIPHER**
GRCh37

 **IRDiRC**
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM



European Journal of Human Genetics

Journal home | Archive | Policy | Full text

Journal home
Advance online publication
1 About EJHG
Current issue
Archive
Practical Genetics

Policy
European Journal of Human Genetics (2013) 26, 721-726; doi:10.1093/hjmg/26.4.691 published online 24 September 2014

International Charter of principles for sharing bio-specimens and data
EMRCopen

Full text
Previous | Next
Table of contents
Download PDF
Send to a friend
View interactive PDF in ReadCube



Impact session

18

- Feedback from the PAC (Virginie Bros-Facer)
- Short update on IRDiRC
- Short update on ERNs (Vicki Hedley)
- Short summary of sustainability session
- Discussion and Wrap-up



Sustainability session - agenda

19

What are the “key assets” that are worth maintaining (relevant for RD research beyond the funding period) and haven’t got sufficient funding in place already? What income would be required, and how could we generate it?

Views on how to sustain RD-Connect post EC funding

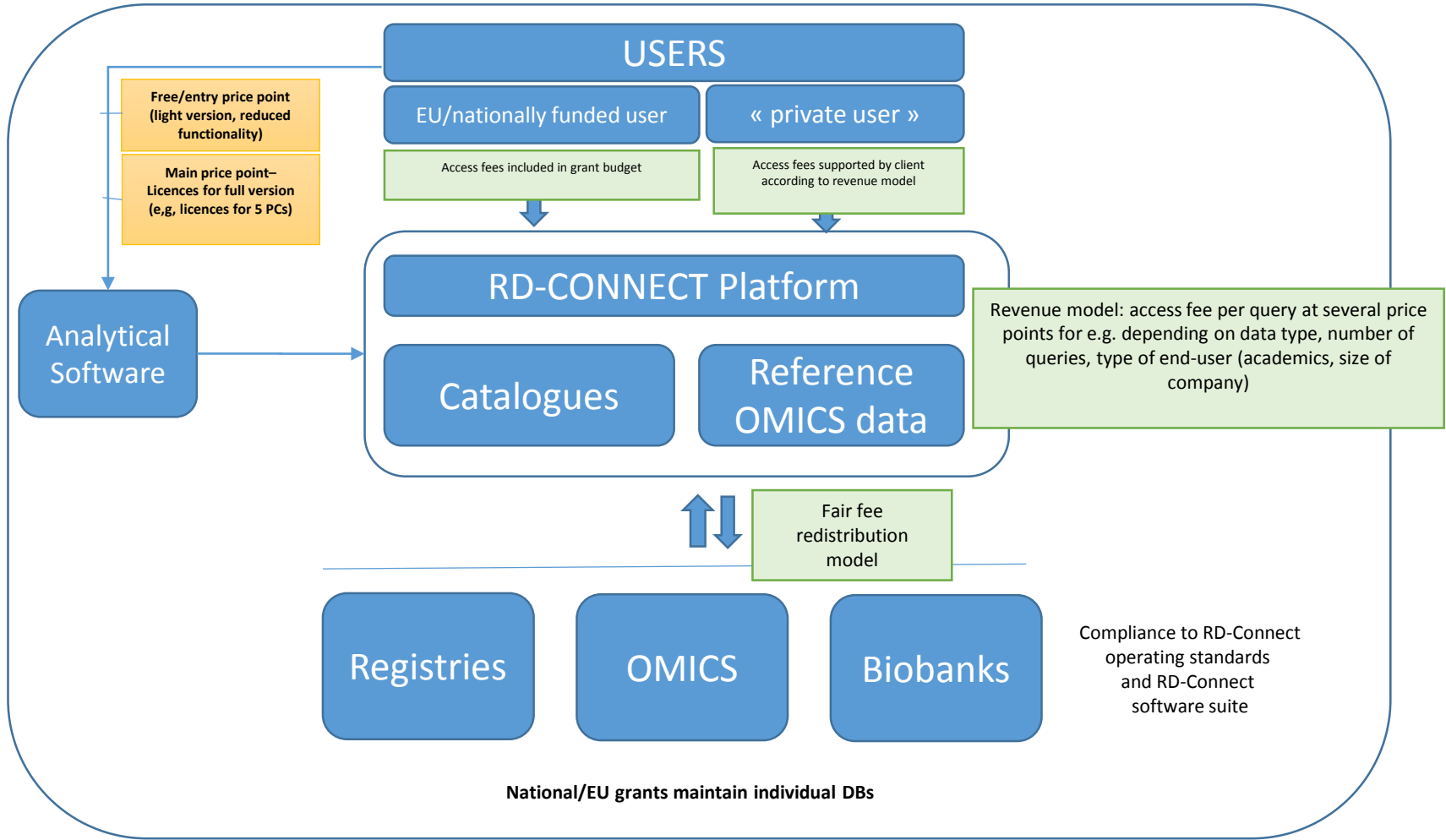
- NeurOmics (Olaf Riess)
- NIH perspective
- Australian perspective
- ELIXIR (Niklas Blomberg)
- BBMRI-ERIC
- ...



Sustainability session – key points

20

- Several key assets identified (platform, knowledge, sample catalogue)
- Are they unique, needed by the user community and what is the cost
- Pro's and con's of keeping the assets together (integration as key value)
- Institutional commitment is important
- ESFRI infrastructures interested (ELIXIR, BBMI)
- Further funding opportunities from the EC (Horizon2020, IMI)
- Potential for commercial activities, fee for service, etc
- Link with other national and international research projects
- Link with European Reference Networks





Next joint project meeting - 2017

22

- Meeting to be held jointly with NeurOmics and EUREnOmics (as their final project meeting)
- Proposed location: Berlin, Germany
- Suggested dates:
 - 24-28 April 2017
 - 2-5 May 2017
 - 8-12 May 2017





What we need you to do next

23

- Keep the good work up
- Take the advice from the SAB on board
- Identify impact and let us know
- Identify the research that gives us success stories within reach
- Identify users and collaborating projects
- Get datasets for the platform from your own institution or department
- Be an ambassador of RD-Connect
- Let us know where you see (funding) gaps in RD research
- Think about sustainability and let us know
- Have a safe trip home!