Integrated European Projects on Omics Research of Rare Diseases

Submitted under FP7 HEALTH.2012.2.1.1-1-B:
Clinical utility of -Omics for better diagnosis of rare diseases
Interaction between the projects

A: Support-IRDiRC

B: NeurOmics

C: RD-Connect

B, B, C representatives attend IRDiRC working groups

Share data, SOPs, stakeholder integration, training, networks, ethics questions, annual meetings

3-monthly calls between coordinators

Share datasets; exchange of scientists
The International Rare Disease Research Consortium (IRDiRC)

- Harmonised research funding initiative launched by the European Union and US NIH – other countries invited to join
- Goals: Diagnosis for all rare diseases and 200 new therapies for RD – by 2020
- Governed by Executive Committee made up of representatives from each member organisation
- Now has 32 committed members from across Europe plus Australia, Canada, USA (+ others joining)
- Each member commits to spending min. 10 million USD over 5 years on research projects contributing to IRDiRC objectives
- Scientific input via 3 scientific committees (diagnostics, therapies, and interdisciplinary) and working groups consisting of experts from funded projects
IRDiRC Governance

Executive Committee

**Interdisciplinary**
- Ethics and governance
- Registries and natural history
- Bio-informatics and data sharing

**Therapeutics**
- Biobanks
- Biomarkers for disease progression and therapy response
- Advanced therapies

**Diagnostics**
- Small molecules
- Ontologies and disease prioritisation
- Model systems

**SCIENTIFIC COMMITTEES**
Approx. 15 members with balanced representation of scientists, patients, industry, etc.

**WORKING GROUPS**
Representatives of funded projects

1 representative per funding body
// 1 representative per group of funders (accumulative funding) //
Representatives of umbrella organisations of patient advocacy groups // Chairs of the Scientific Committees
Neuromics

Integrated European Project on Omics
Research of Rare Neuromuscular and Neurodegenerative Diseases

5 year project
started 1st October 2012

Coordinator: Olaf Riess, University of Tübingen
Co-coordinator: Brunhilde Wirth, University of Cologne
Gert-Jan von Ommen, Leiden University
Diseases and disease groups

Sarah Tabrizi
Alexis Brice
Ludger Schoels
Thomas Klockgether
Brunhilde Wirth
Vincent Timmermann
Hanns Lochmüller
Francesco Muntoni
Gert-Jan van Ommen
Mike Hanna
Main impact

**Main deliverables**
- Genes, modifier, biomarkers, diagnostic panels, targets, interventions

**Main impacts**
- NEUROMICS clinical infrastructure
  - Network, cohorts, registries, biobanks, trial-readiness
- Diagnostic tools
- Comprehensive knowledge base
- Diagnostic & therapeutic biomarkers
- Therapeutic interventions

**Transfer to clinical utility**

**Improve patient health**
- Improved diagnosis
- Personalised treatments
- Improved QoL

**Social impact**
- Reduced health costs
- Improved QoL
- Impact on policies (IRDiRC)

**Transfer to further NDD/NMD/RD**
- Pathogenic commonalities
- Shared phenotypical and omics markers as well as targets

**Economic gain**
- Diagnostics market
- Therapeutics market
- Biomarker market
- Sequencing market

ESHG Meeting | June 8-11 2013 | Paris (France)
High-throughput research for rare kidney diseases

Franz Schaefer
University of Heidelberg
Targeted Rare Kidney Disorders

WP2/3: Nephrotic glomerulopathies

WP5: Complement-mediated nephropathies

WP4: Tubulopathies

WP6: Kidney malformations
26 academic research groups
19 institutions in 11 countries
8 industry partners
Cohorts and Biorepositories

Technologies
- Exome sequencing
- Transcriptomics: Expression profiling, ChipSeq
- miRNAomics
- Proteomics
- Metabolomics
- Epitope screening

WP7
- Multilevel integrative bio-informatics
- ’Functiomics’: Cell based assays, ipSC, Zebrafish, Xenopus, KO/KI mice

Targeted Output
- Phenotype standardization
- Reference -omics profiles
- Drug candidate compounds
- Models to support trials

Molecular disease ontologies
- Rapid diagnostic tests
- Novel biomarkers
- Sustainable resources

IRDiRC Goals: Personalized Prediction, Diagnosis and Therapies for Patients with Rare Diseases
An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research

Overarching objectives:

- Development of an integrated, quality-assured and comprehensive platform in which complete clinical profiles are combined with -omics data and sample availability for rare disease research, in particular IRDiRC-funded research.
RD-Connect workpackage overview

WP1: Coordination
Hanns Lochmüller
Newcastle and TREAT-NMD

WP2: Patient registries
Domenica Taruscio
ISS and EPIRARE

WP3: Biobanks
Lucia Monaco
Fondaz. Telethon & EuroBioBank

WP4: Bioinformatics
Christophe Béroud
INSERM Marseille

WP5: Unified platform
Ivo Gut
CNAG Barcelona

WP6 Ethical/legal/social
Mats Hansson
Uppsala

WP7: Impact and innovation
Kate Bushby
Newcastle and EUCERD/ EJARD
Patient registries: developing best practice for registries used for research – establishing interoperability standards, common data elements => feeding into central platform

Biobanks: developing interoperability standards, common MTAs, searchable online catalogue of sample availability (building on EuroBioBank and BBMRI) => feeding into central platform

Bioinformatics tools: developing and integrating clinical bioinformatics tools and making them accessible through the central platform and via APIs and web services

Ethical, legal and social issues: addressing data sharing and informed consent for omics research, proposing a regulatory framework for linking RD medical and personal data, integrating patient perspective
Sitges (Barcelona), 25 January 2013