Integrated European Projects on Omics Research of Rare Diseases

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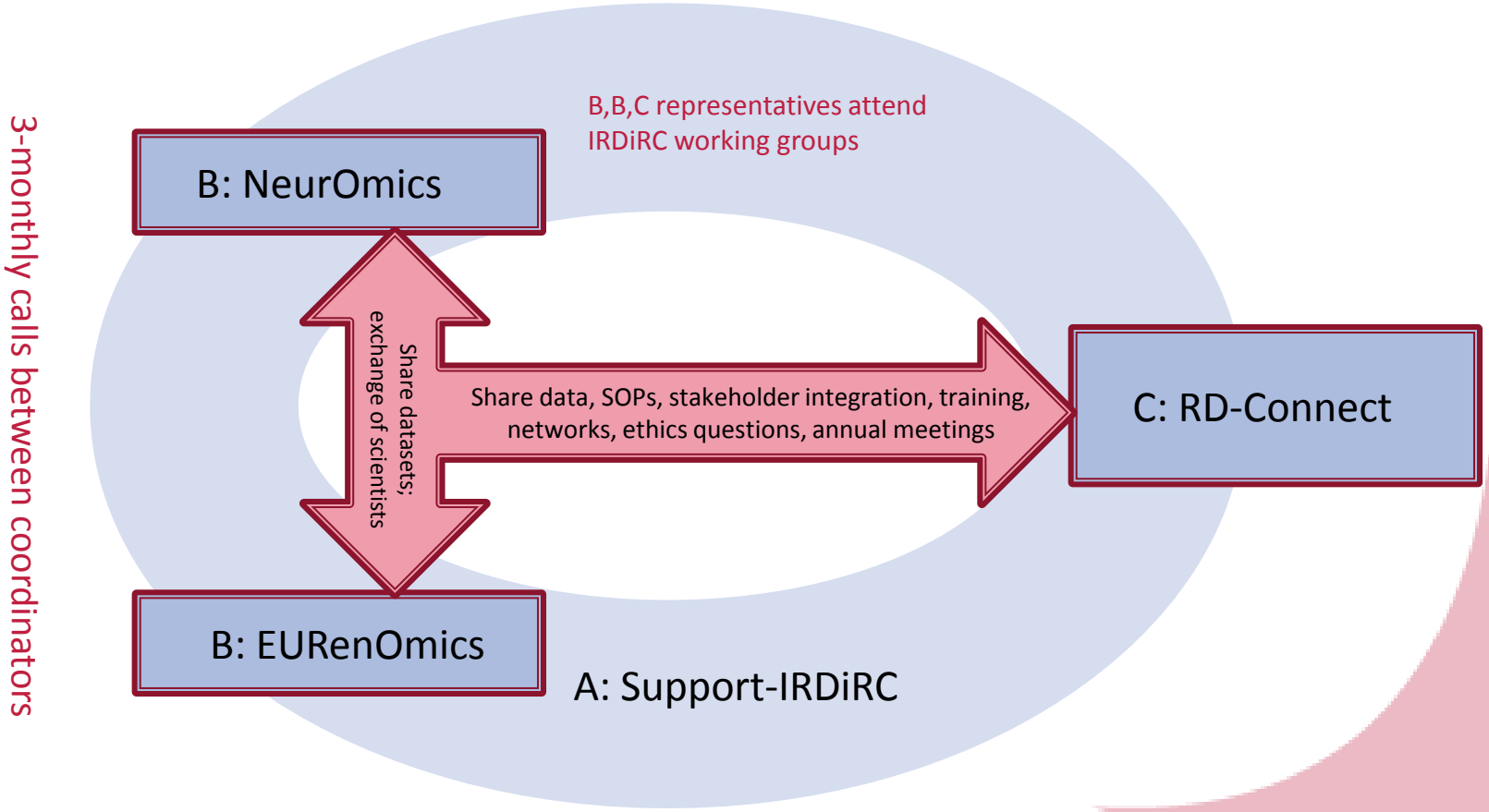
Clinical utility of -Omics for better diagnosis of rare diseases

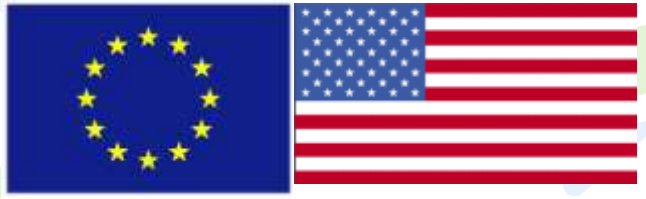
Olaf Riess (Universität Tübingen)

Interaction between the projects



IRDiRC – the International Rare Disease Research Consortium

- 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

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

Executive Committee

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

Interdisciplinary	Therapeutics	Diagnostics

WORKING GROUPS
Representatives of funded projects

Ethics and governance	Biobanks	Biomarkers for disease progression and therapy response	Small molecules	Ontologies and disease prioritisation	Sequencing
Registries and natural history	Bio-informatics and data sharing	Advanced therapies	Regulatory	Model systems	Genome / Phenome

RD Connect

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

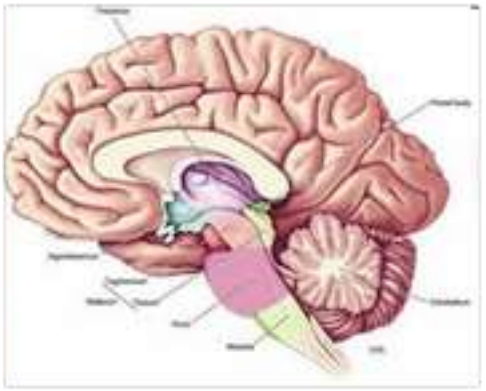
Hanns Lochmüller

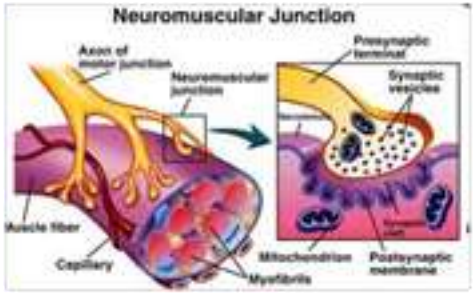
Francesco Muntoni

Gert-Jan van Ommen

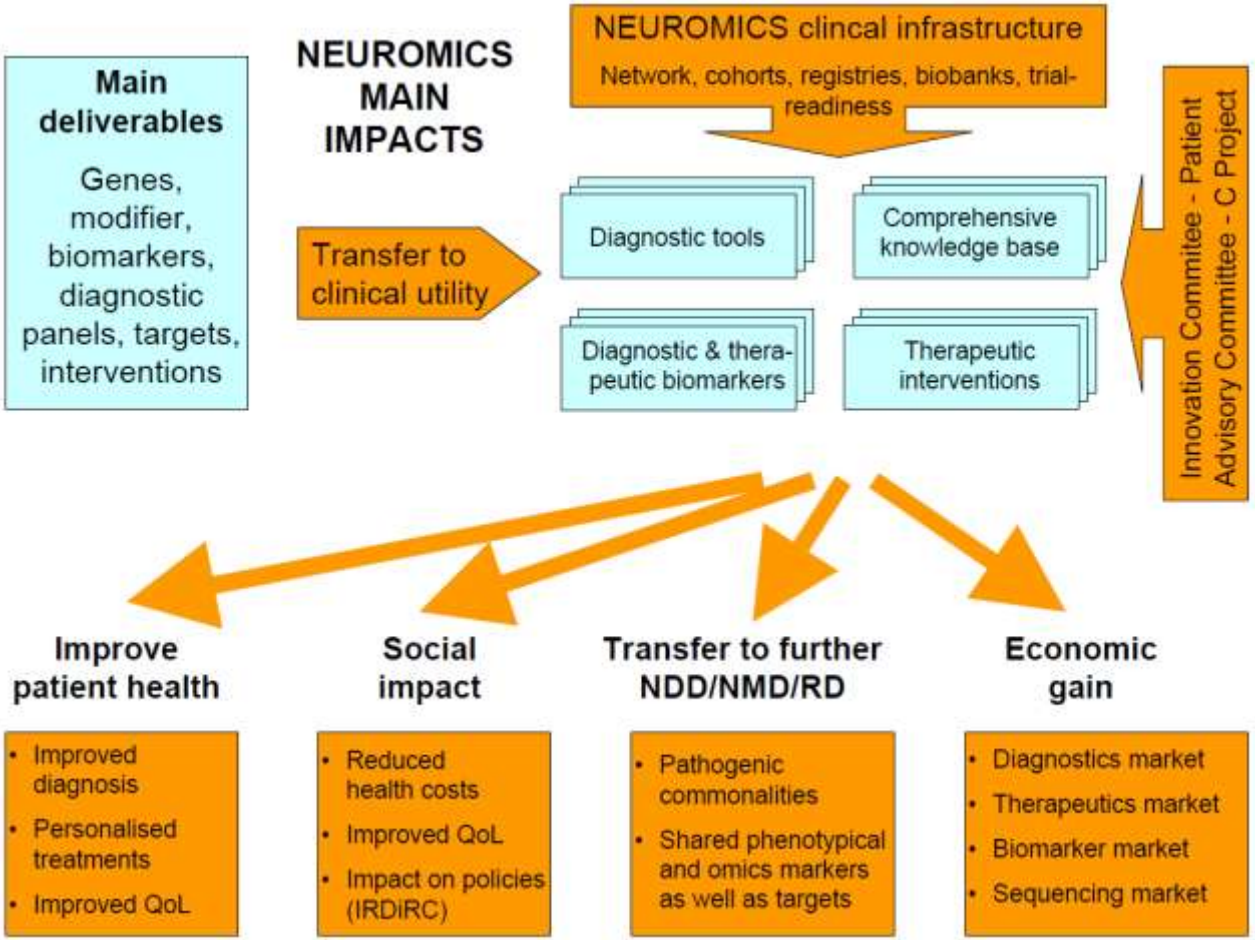
Mike Hanna

- Huntington disease (HD)
- Fronto-temportal lob dementia (FTLD)
- Hereditery spastic paraplegia (HSP)
- Ataxias (ADCA, ARCA, CA)
- Spinal muscular atrophies & lower motoneuron diseases (SMA, LMND)
- Hereditary motor neuropathies (HMN)
- Congenital myastenic syndrome (CMS)
- Congenital dystrophies & myopathies (CMD, CMY)
- Muscular dystrophies (DMD, BMD, FSHD, LGMD)
- Muscular channelopathies (MCP)





Main impact


Main deliverables
Genes, modifier, biomarkers, diagnostic panels, targets, interventions
NEUROMICS MAIN IMPACTS
Transfer to clinical utility
NEUROMICS clincal infrastructure
Network, cohorts, registries, biobanks, trial-readiness
Diagnostic tools
Comprehensive knowledge base
Diagnostic & thera-peutic biomarkers
Therapeutic interventions
Innovation Commitee - Patient Advisory Committee - C Project
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

EUReni Omics

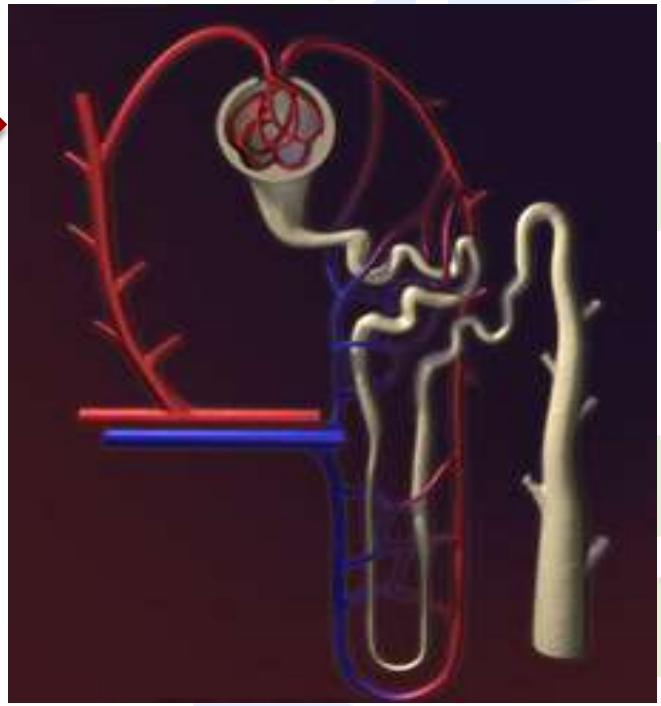
High-throughput research for rare kidney diseases

Franz Schaefer
University of Heidelberg

Targeted Rare Kidney Disorders

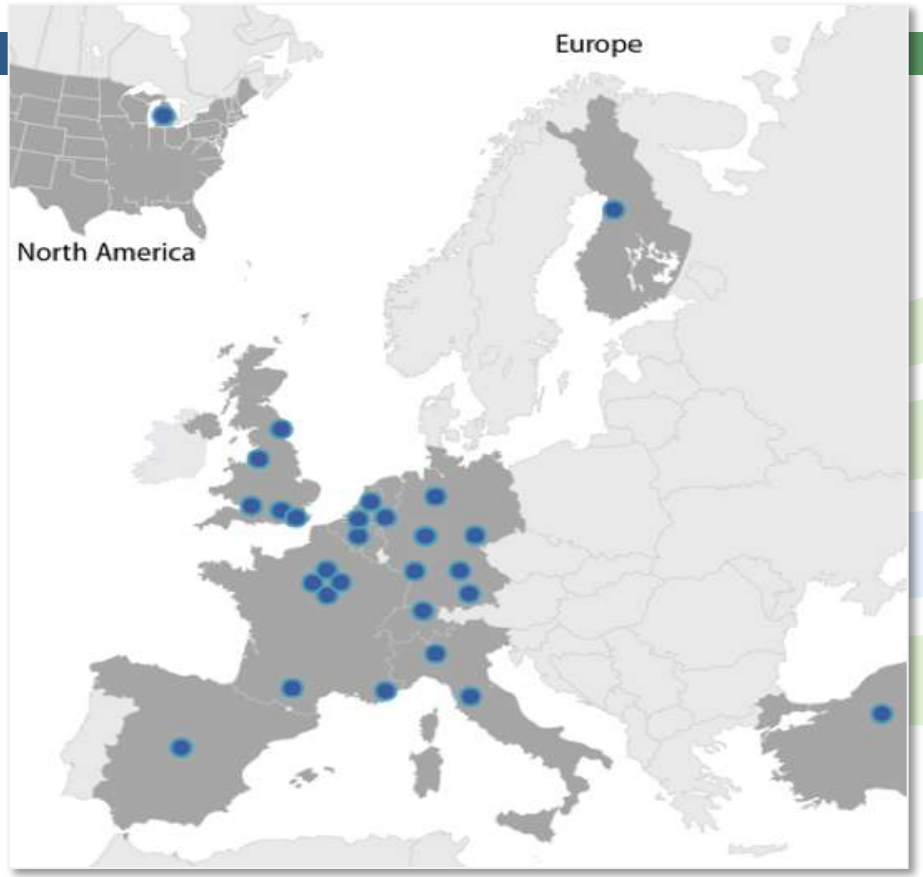
WP2/3:
Nephrotic glomerulopathies

WP5:
Complement-mediated nephropathies

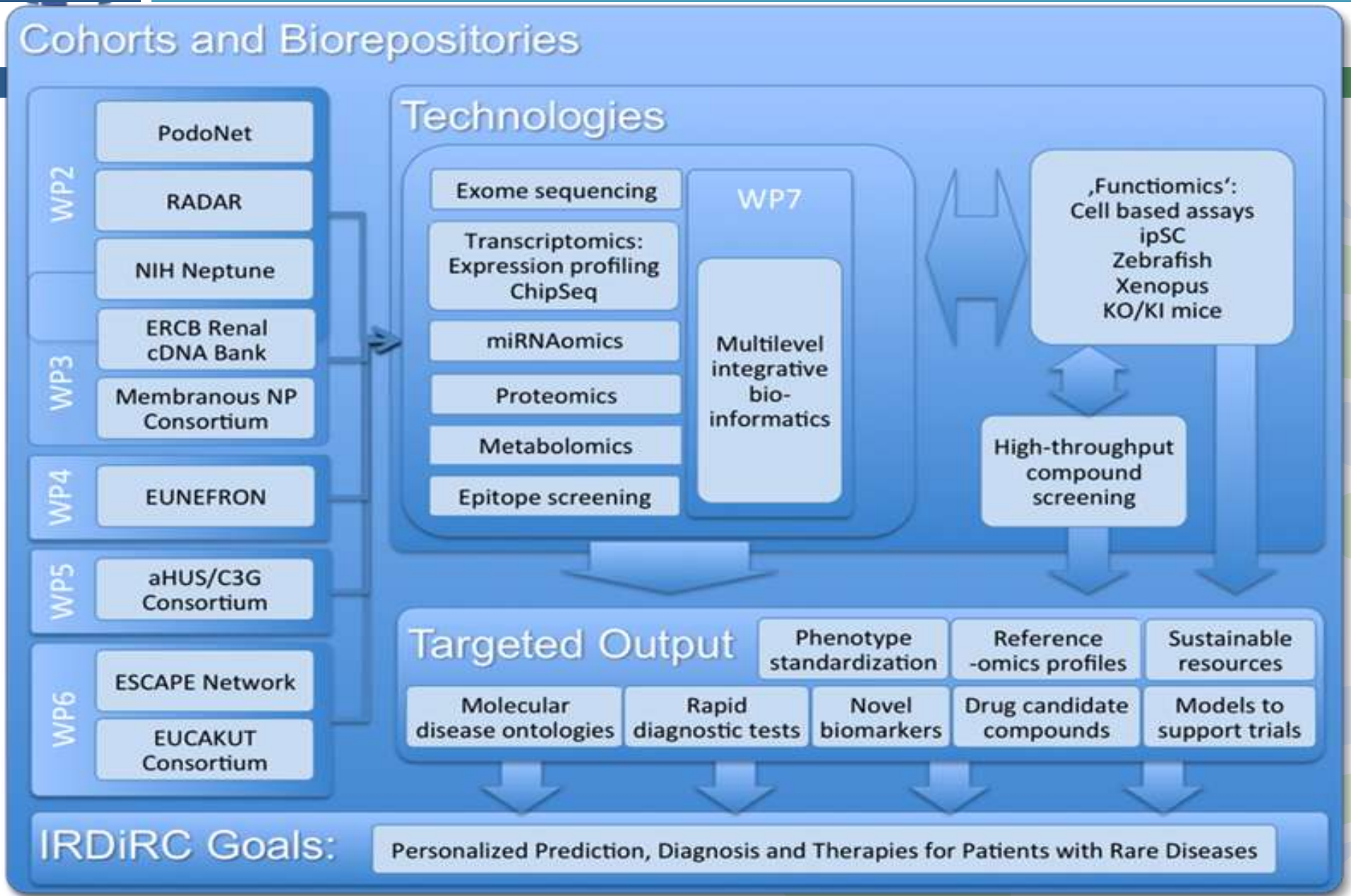


WP4:
Tubulopathies

WP6: Kidney malformations



26 academic research groups
19 institutions in 11 countries
8 industry partners


Cohorts and Biorepositories
WP2
PodoNet
RADAR
NIH Neptune
WP3
ERCB Renal cDNA Bank
Membranous NP Consortium
WP4
EUNEFRON
WP5
aHUS/C3G Consortium
WP6
ESCAPE Network
EUCAKUT Consortium
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
High-throughput compound screening
Targeted Output
Phenotype standardization
Reference -omics profiles
Sustainable resources
Molecular disease ontologies
Rapid diagnostic tests
Novel biomarkers
Drug candidate compounds
Models to support trials
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

RD Connect

RD-Connect additional objectives

- **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