



IRDiRC

INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

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Office of Rare Diseases Research (ORDR)
National Center for Advancing Translational Science (NCATS)
National Institutes of Health (NIH)
Department of Health and Human Services
Best Practices in Clinical Study Design for Rare Diseases
The George Washington University
Washington, DC
April 30, 2013**

International Rare Diseases Research Consortium

- **Co-operation At International Level
To Stimulate, Better Coordinate & Maximise Output
Of Rare Disease Research Efforts Around The World**
- **Establish and Link Global Rare Diseases Community:
Research Investigators, Protocols and Clinical Trials,
Academia, Industry, Government, and PAG, and Regulatory
Partnerships**
- **<http://www.irdirc.org/>**

IRDIRC Governance Document

- **Defines the aims of the consortium; mandate and composition of committees and working groups; rules for procedures, nomination of members, conflicts of interest etc.**

IRDIRC Policies and Guidelines

- **Will establish a common policy framework addressing various aspects of research, such as, sharing of data and samples, common quality standards, interoperability and harmonisation of ontologies and bio-banks, rapid release of research results, etc.**

IRDiRC Vision And 2020 Goals In Rare Diseases Research



200 New Therapies



Means to Diagnose Most Rare Diseases

32 committed members

Europe

E-RARE Consortium (EU)
European Commission (EU)
EURORDIS (EU)
French Association against Myopathies (FR)
French National Research Agency (FR)
German Federal Ministry of Education and research (DE)
Italian Higher Institute of Health Research (IT)
Italian Telethon Foundation (IT)
Lysogene (FR)
Netherlands Organisation for Health Research and Development
Prosensa (NL)
Spanish Carlos III Health Institute (ES)
UK National Institute for Health Research (UK)



Asia

BGI (CN)

Australia

Western Australian Department of Health (AU)

North America

Canadian Institutes for Health Research (CA)
FDA Office of Orphan Products Development (US)
Genome Canada (CA)
Genetic Alliance (US)
NHGRI Mendelian Disorders Genome Centres (US)
National Center for Advancing Translational Sciences (US)
National Cancer Institute (US)
National Institute of Neurological Disorders and Stroke (US)
National Institute of Arthritis and Musculoskeletal and Skin Diseases (US)
National Institute of Child Health and Human Development (US)
National Eye Institute (US)
NKT Therapeutics (US)
NORD (US)
Office of Rare Diseases Research NCATS (US)
PTC Therapeutics (US)
Sanford Research (US)
Shire (US)

IRDiRC Governance Structure



EU-funded SUPPORT-IRDiRC provides organisational support for Scientific Committees and Working Groups

How Does It Work?

- **Identify And Define Shared Strategic Goals**
- **Agree To Approach Jointly And Pool Resources**
- **Let Each Organisation Use Its Own Funding Mechanisms/Timing**
- **Agree To Share Data / Standards**

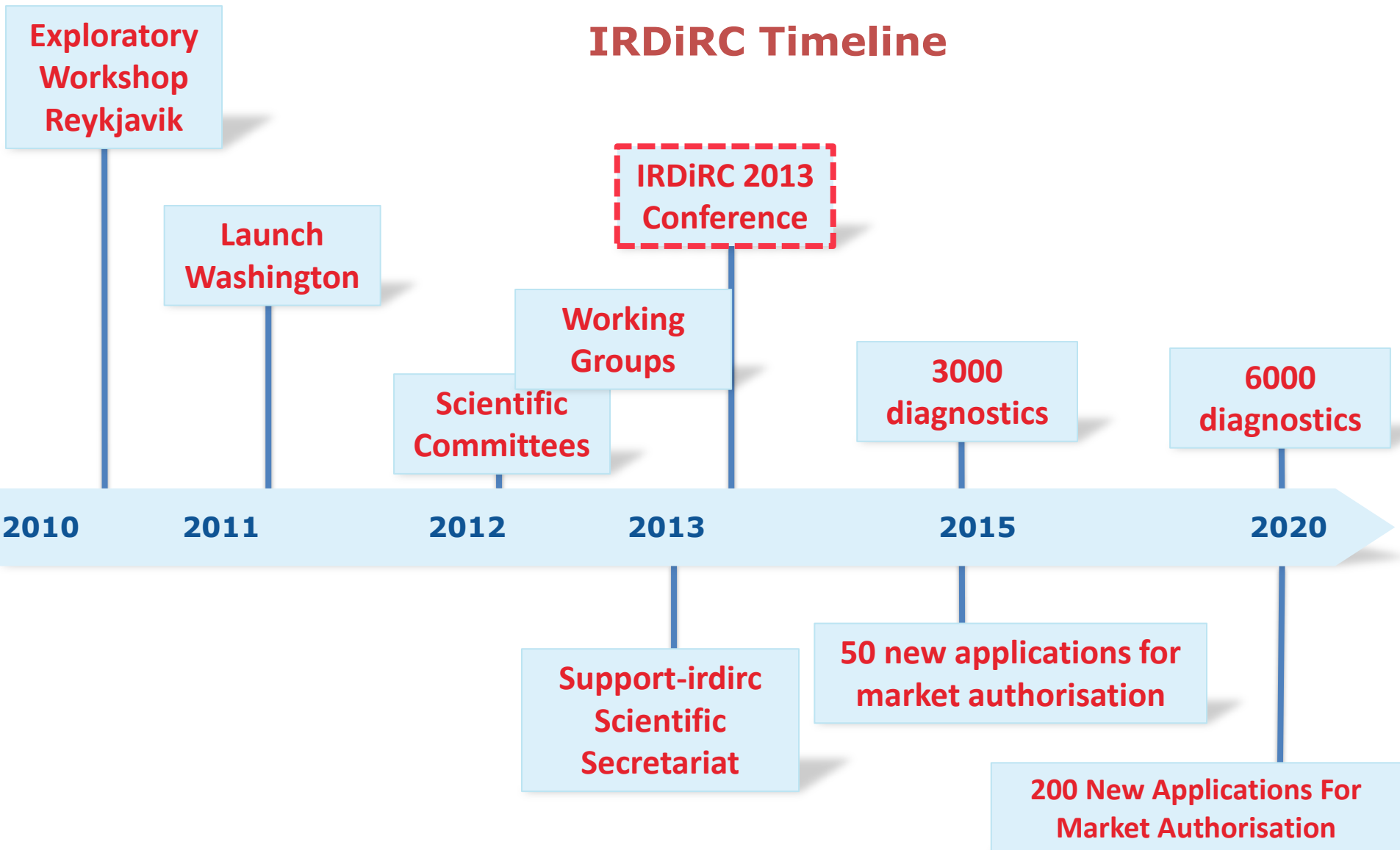


Alignment – Flexibility - Commitment

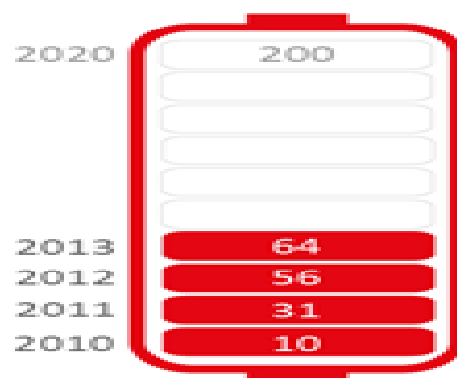
Working Groups To Advise The Scientific Committees

- Ontologies
- Sequencing
- Model Systems
- Genomics
- Bioinformatics
- Bio-banks
- Registries
- Biomarkers
- Ethics
- Repurposing Drugs
- Therapies
- Regulatory Issues

IRDiRC Timeline



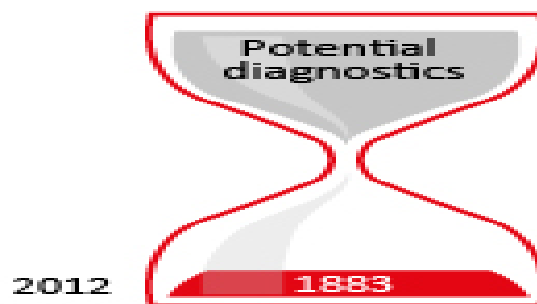
NEW THERAPIES



Objective 2020: 200 new therapies

Disclaimer: the numbers do not reflect IRDiRC initiatives only

NEW DIAGNOSTICS



Objective 2020: diagnostics for all rare diseases

Disclaimer: the numbers do not reflect IRDiRC initiatives only

Contact Information

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E-mail: ORDR@nih.gov

Web Site : <http://rarediseases.info.nih.gov/>

Genetic and Rare Diseases Information Center

Toll-free: 888-205-2311 TTY: 888-205-3223

E-mail: GARDinfo@nih.gov

Web site: <http://rarediseases.info.nih.gov/GARD/>

