



Rare disease research in FP7, Outlook in Horizon 2020, IRDIRC

ECRD, Brussels, 24 May 2012

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International Rare Diseases Research Consortium (IRDIRC)

Co-operation at international level
to stimulate, better coordinate & maximise output
of rare disease research efforts around the world



Innovation



IRDIRC – basic principles

- Teams up public and private organisations investing in rare diseases research
- Research funders with relevant programmes >\$10 million US over a 5-year period can join & work together
- Each organisation funds research its own way
- Funded projects adhere to a common framework

Current 25 EXEC members



Europe

European Commission

German Federal Ministry of Education and research

Italian Higher Institute of Health Research

Italian Telethon Foundation

French Association against Myopathies

French National Research Agency

Netherlands Organisation for Health Research and Development

Lysogene (FR)

Prosensa (NL)

Spanish Carlos III Health Institute

UK National Institute for Health Research

Shire (IE)



Australia

Western Australian Department of Health

Research and

North America

Canadian Institutes for Health Research (CA)

Genome Canada (CA)

Sanford Research (US)

Mendelian Disorders Genome Centres(US)

National Centre for Translational Therapeutics (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)

Office of Rare Diseases (US)

Food and Drug Administration (US)

IRDIRC governance

- **Executive Committee (EXEC)**
Composed of representatives from IRDiRC funding bodies; coordinates research funding and nominates Scientific Committee members, approves policies and guidelines
- **Scientific Committees**
Each with about 15 members to represent stakeholders in a balanced way; propose research priorities, policies and guidelines to the EXEC, assess progress, and agrees on procedures and standards..
- **Working groups**
Represent all IRDiRC projects; ensure synergies, propose standards for best use of data and results

IRDiRC timeline



**Launch of
IRDiRC**

**Scientific
Committees**



3000 diagnostics



6000 diagnostics



YEAR

2012

2015

2020

**Working
Groups**

**50 new applications for
market authorisation**

**200 new applications for
market authorisation**





Rare diseases research

History of support at the European level

EU has invested in research on rare diseases for more than 2 decades

FP5 (1998-2002): 47 projects funded, € 64 million in total

ftp://ftp.cordis.europa.eu/pub/lifescihealth/docs/reprint_rec48300_rare_dis_060207.pdf

FP6 (2002-2006): 59 projects funded, € 230 million in total

<http://cordis.europa.eu/lifescihealth/major/rare-diseases-projects-1.htm>



EC support to rare diseases research FP7 Health Theme 2007-2013

66 ongoing projects: EC support around € 325 million

- Europe-wide studies of **natural history** and **pathophysiology**: development of in vitro/in vivo models, registries and biobanks, identification of biomarkers etc.
- Development of **preventative, diagnostic and therapeutic interventions**, including pharmacological approaches and innovative approaches such as cell and gene therapies, and regenerative medicine.
- In **most diseases areas**: neurology, immunology, cancer, pneumology, dermatology, uro-gynaecology, metabolism etc.



Work Programme 2012 for Health Theme

€ 108 million earmarked for the following topics:

Support for international rare diseases research

Clinical utility of -omics for better diagnosis of rare diseases

Databases, biobanks and clinical 'bio-informatics' hub for rare diseases

Preclinical and/or clinical development of substances with a clear potential as orphan drugs

Observational trials in rare diseases

Best practice and knowledge sharing in the clinical management of rare diseases



NEXT CALL FOR PROPOSALS EXPECTED TO BE OPENED IN JULY 2012

**FP7 Health - 2013 orientation paper
available on Cordis:**

http://cordis.europa.eu/fp7/health/home_en.html

Indicative topics related to rare diseases:

- HEALTH.2013.1.2-1: Development of imaging technologies for therapeutic interventions in rare diseases
Min. 30 % EU Contribution to SME(s)
- HEALTH.2013.4.2-3: New methodologies for clinical trials for small population groups
 - statistical design methodologies for clinical trials in particular for rare diseases or personalised medicine

Based on working document, refer to final published call text

Other potential entry points for rare disease research:

- HEALTH.2013.0-1: Boosting the translation of FP projects' results into innovative applications for health
Min. 50 % EU Contribution to SME(s)
- HEALTH.2013.2.1.1-1: Functional validation in animal and cellular models of genetic determinants of diseases and ageing processes
Min. 30 % EU Contribution to SME(s)
- HEALTH.2013.1.4-1. Controlling differentiation and proliferation in human stem cells intended for therapeutic use
Min. 15 % EU Contribution to SME(s)

Based on working document, refer to final published call text



Horizon 2020: The next Framework Programme for research and innovation

Proposed budget: €80bn, a 46% increase compared to FP7

Europe needs cutting edge research and innovation

Essential to ensure competitiveness, growth and jobs

Vital to tackle pressing societal challenges

3% of GDP invested in R&D: headline target of Europe 2020



Innovation



Horizon 2020: Health, Demographic Change and Wellbeing Challenge – proposal ~ €8.5bn

Understanding the determinants of health (including environmental and climate related factors), improving health promotion and disease prevention;

Developing effective screening programmes and improving the assessment of disease susceptibility

Improving surveillance and preparedness

Understanding disease

Developing better preventive vaccines

Improving diagnosis

Using in-silico medicine for improving disease management and prediction

Treating disease

Transferring knowledge to clinical practice and scalable innovation actions

Better use of health data

Improving scientific tools and methods to support policy making and regulatory needs

Active ageing, independent and assisted living

Individual empowerment for self-management of health

Promoting integrated care

Optimising the efficiency and effectiveness of healthcare systems and reducing inequalities through evidence based decision making and dissemination of best practice, and innovative technologies and approaches.



Further information

EU research

<http://ec.europa.eu/research>

EU Health Research

http://ec.europa.eu/research/health/index_en.html

Seventh Framework Programme

<http://ec.europa.eu/research/fp7>

FP7 Rare diseases

http://ec.europa.eu/research/health/medical-research/rare-diseases/index_en.html

Information on research programmes and projects

<http://cordis.europa.eu/>

Information requests

<http://ec.europa.eu/research/enquiries/>