



**EURODIS**  
Rare Diseases Europe



# HORIZON 2020 AND RARE DISEASES UPCOMING OPPORTUNITIES FOR RESEARCH ON RARE DISEASES

## INFORMATION SESSION



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30<sup>TH</sup> OCTOBER 2013

1. FP7 and its predecessors: how **past EU research programmes** contributed to RD research
2. What do we know about **HORIZON 2020**
3. International collaboration on RD research: **IRDiRC**, an *ad hoc* consortium

*“Rare disease research is revealing Nature’s secrets, held for millennia, with unexpected benefits”*

William A. Gahl

1. FP7 and its predecessors: how **past EU research programmes** contributed to RD research

## → Role of European Commission in research :

1. **Funding agency:** competitiveness of EU industry, support in predefined areas
  2. **Preparing and implementing policies**
  3. **Bringing down barriers**
    - between countries:
      - multinational consortia and teams
      - researchers from any country in the world can participate
      - coordination among national funding programmes
    - between different types of organisations: universities, research centres, SMEs, large companies, etc.
    - between disciplines: increased focus on translational research
  5. **Encourage mobility:** Marie Curie fellowships available for researchers and for host institutes
  6. **Supporting common infrastructures**
- **main instrument = Framework Programmes (FP)**



	<b>FP5</b> <b>[1998-2002]</b>	<b>FP6</b> <b>[2002-2006]</b>	<b>FP7</b> <b>[2007-2013]</b>	<b>HORIZON 2020</b> <b>[2014-2020]</b>
<b>Budget for the entire Research Framework Programme</b>	14 960 M€	17 500 M€	54 582 M€	70 200 M€
<b>Health Specific Programme</b>	“Improving the quality of life and management of living resources”	“Life sciences, genomics and biotechnology for health”	Health Theme  (one of the 10 Themes of the ‘Cooperation Programme’)	“Health, Demographic Change and Wellbeing ”  (part of the broader area “Societal challenges”)
<b>Budget of the ‘Health’ programme</b>	2 413 M€	2 514 M€	6 100 M€	7 400 M€
<b>Total budget allocated to RD research</b>	<b>64 M€</b>	<b>230 M€</b>	<b>530 M€</b>	<b>?</b>
<b>n° RD specific funded projects</b>	<b>47</b>	<b>59</b> also including an ERA-Net project (E-Rare) and one SiS (CAPOIRA)	<b>103</b>	<b>?</b>

**FOCUS:** was on:

- Europe-wide studies of natural history, pathophysiology and
- development of preventive, diagnostic and therapeutic interventions.

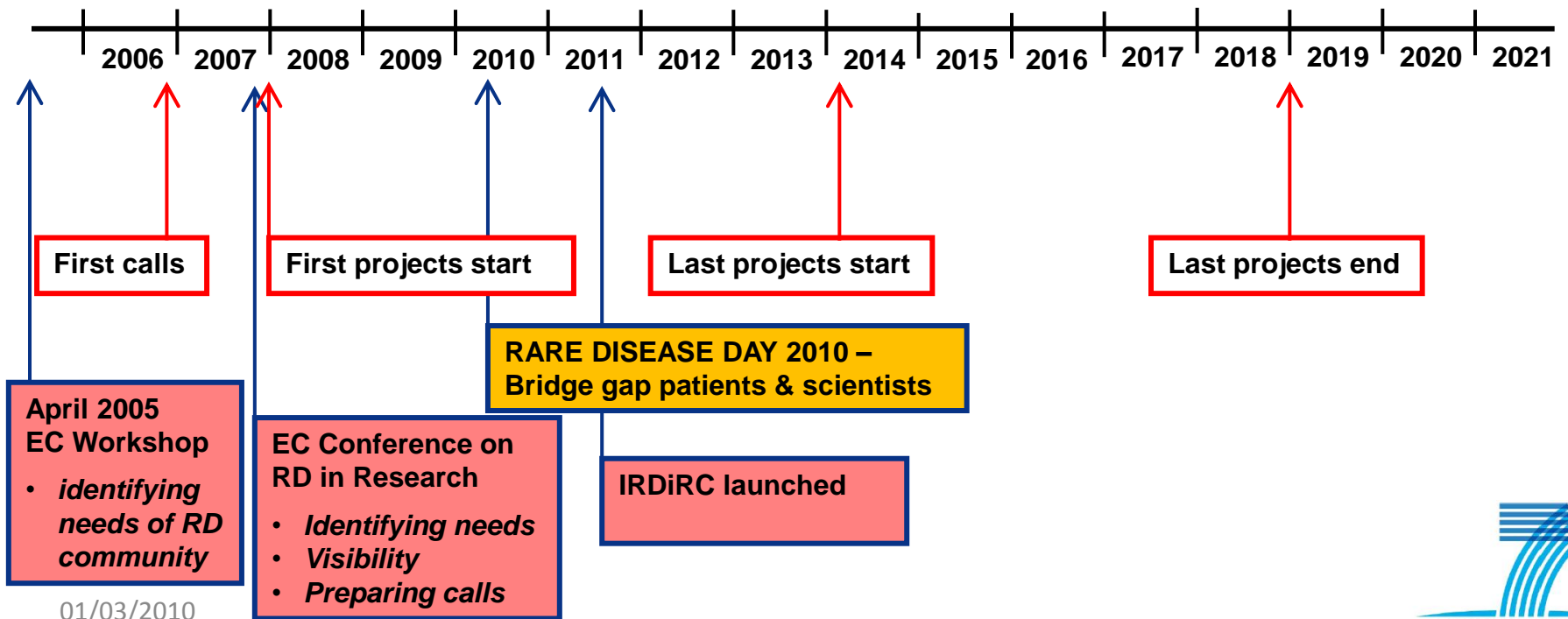
**EXPECTED IMPACT:** help identifying and mobilising the critical mass of expertise in order to:

- shed light on the **course and/or mechanisms of rare diseases**, or
- test **preventative, diagnostic and/or therapeutic approaches** to alleviate the negative impact of the disease on the quality of life of the patients and their families, as appropriate, depending on the level of knowledge concerning the specific (group of) disease(s) under study.

# Timeframe for FP7 projects

Projects resulting from FP7 calls operating

Official duration of FP7 2007-2013



01/03/2010

# FP7 AND RD RESEARCH – FIGURES

**TOTAL BUDGET:**

**530 million €**

**n° PROJECTS:**

**103**

**Specific RD area « 2.4.4 »  
or specific topics RD-related:**

**260,5 million €**

**Other areas where RD-related  
projects have been funded:**

**270 million € approx (tbc)**



	<b>Specific RD area « 2.4.4 » or specific topics RD-related:</b>	<b>n° projects</b>	<b>total budget (M€)</b>
<b>2007</b>	HEALTH-2007-2.4.4-1: <b>Natural course and patho-physiology of rare diseases</b>	10	29
	HEALTH-2007-2.4.4-2: <b>Research capacity-building in the field of rare diseases</b>	1	1
<b>2009</b>	HEALTH-2009-2.4.4-1: <b>Rare neurological diseases</b>	7	41
	HEALTH-2009-2.4.4-2: <b>Preclinical development of substances with a clear potential as ODs</b>	2	4
<b>2010</b>	HEALTH-2010-2.4.4-1: <b>Clinical development of substances with a clear potential as ODs</b>	6	31
	HEALTH-2010-2.4.4-2: <b>ERA-NET for rare diseases</b>	1	2
<b>2012</b>	HEALTH-2012-2.4.4-1: <b>Preclinical and clinical development of substances with clear potential as ODs</b>	10	57
	HEALTH-2012-2.4.4-2: <b>Observational studies</b>	5	15
	HEALTH-2012-2.4.4-3: <b>Best practice, knowledge sharing in the clinical management of RD</b>	1	2
	HEALTH-2012-2.1.1-1: <b>-omics for RDs</b>	4	38
<b>2013</b>	HEALTH.2013.4.2-3: <b>New methodologies for clinical trials for small population groups</b>	2	6
	HEALTH.2013.1.2-1: <b>Imaging technologies for therapeutic interventions in RD</b>	6	34,5
		<b>55</b>	<b>260,5</b>

# Rare diseases: 2007 call for proposals

## HEALTH-2007-2.4.4-1: Natural course and patho-physiology of rare diseases

→ 10 projects selected for funding - total budget **€ 29 million**

- PEMPHIGUS - **Pemphigus** - From autoimmunity to disease
- EURO-PADNET - The pathophysiology and natural course of patients with **Primary Antibody Deficiencies (PAD)**
- CUREHLH - European initiative to improve knowledge, treatment and survival of **haemophagocytic syndromes** in children
- EURADRENAL - Pathophysiology and natural course of **autoimmune adrenal failure**
- EUCLYD - European Consortium for **Lysosomal Disorders**
- EURIPFNET - European IPF Network: natural course, patho-mechanisms & novel treatments in **Idiopathic Pulmonary Fibrosis**
- EUNEFRON- European Network for **Orphan Nephropathies**

# Rare diseases: 2007 call for proposals

HEALTH-2007-2.4.4-2: **Research capacity-building in the field of rare diseases**

→ 1 project selected for funding - budget of **€ 1 million**

- RD PLATFORM - European Platform of **Integrated Information Services for Researchers** in the Field of Rare Diseases and Orphan Drugs Supporting Team and Project Building

# Rare diseases: 2009 call for proposals

## HEALTH-2009-2.4.4-1: Rare neurological diseases

→ 7 projects selected for funding – total budget € 41 million

- BIO-NMD - Identifying and validating pre-clinical biomarkers for diagnostics and therapeutics of **Neuromuscular Disorders**
- FIGHT-MG - **Myasthenias**, a group of immune mediated neurological diseases: from aetiology to therapy
- Fighting blindness of **Usher syndrome**: diagnosis, pathogenesis and retinal treatment
- LeukoTreat - Therapeutic challenge in **Leukodystrophies**: Translational and ethical research towards clinical trials
- MEFOPA- European Project on Mendelian forms of **Parkinson's Disease**

# Rare diseases: 2009 call for proposals

HEALTH-2009-2.4.4-2: Preclinical development of substances with a clear potential as orphan drugs

→ 2 projects selected for funding – total budget **€ 4 million**

- PRATH - Preclinical study of recombinant human anti-C5 for the treatment of **Atypical Haemolytic Uraemic syndrome**
- RDVCF - Rod-derived **Cone Viability Factor**

# Rare diseases: 2010 call for proposals

HEALTH-2010-2.4.4-1: **Clinical development of substances with a clear potential as orphan drugs**

→ 6 projects selected for funding for a budget of **€ 31 million**

- IMPACTT - Immunoglobulin IgY pseudomonas. A clinical trial for **cystic fibrosis** treatment
- AIPGENE - Novel Gene therapy for **Acute Intermittent Porphyria**
- MABSOT - Treatment of Delayed Graft Function post solid organ transplantation
- GENEGRAFT - Phase I/II ex vivo gene therapy clinical trial for **recessive dystrophic epidermolysis bullosa** using skin equivalent grafts genetically corrected with a COL7A1-encoding SIN retroviral vector
- PADDINGTON - Pharmacodynamic Approaches to Demonstration of Disease-Modification in **Huntington's Disease**
- ALPHA-MAN - Enzyme Replacement Therapy in **alpha-Mannosidosis**

# Rare diseases: 2010 call for proposals

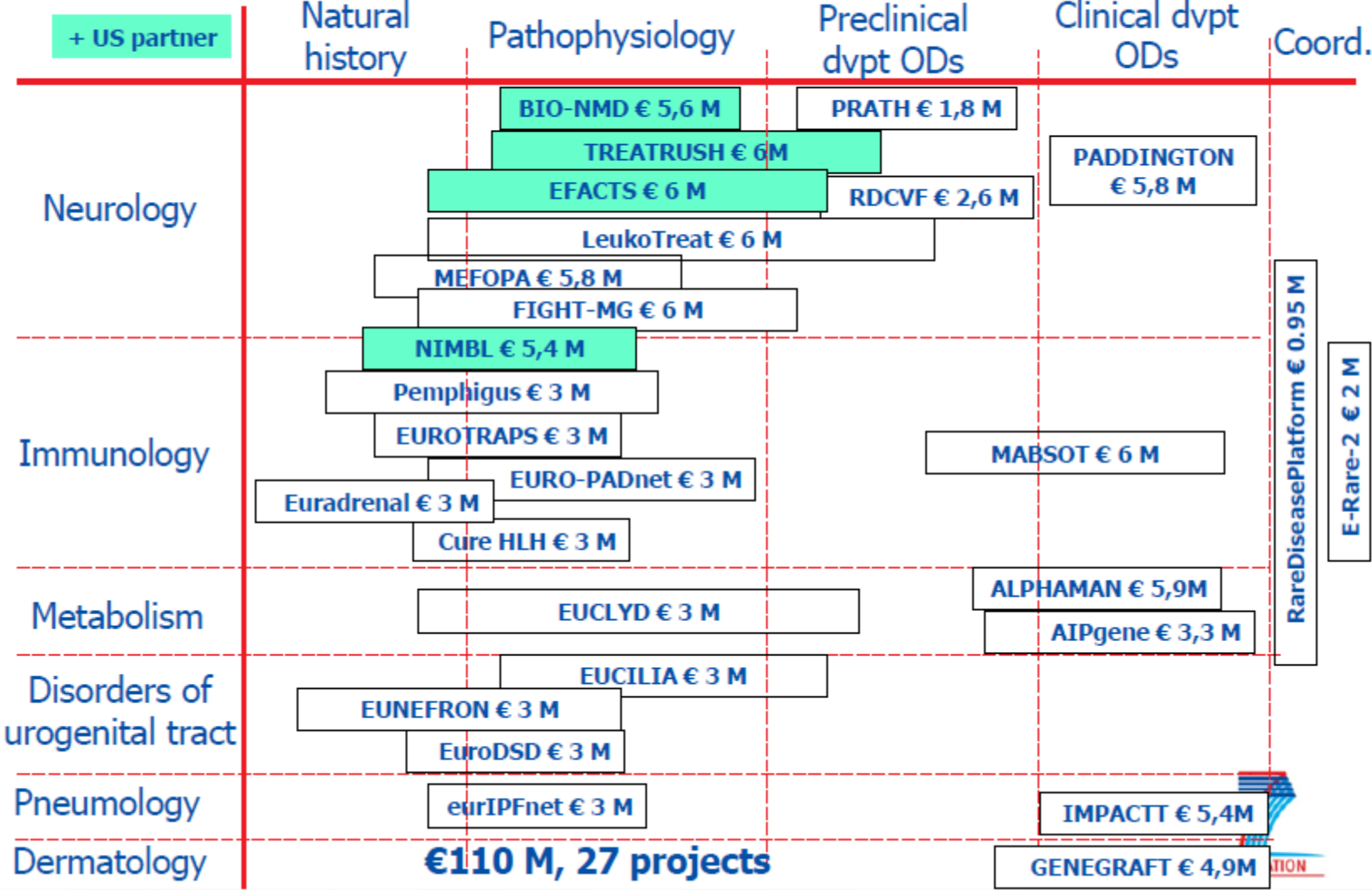
HEALTH-2010-2.4.4-2: ERA-NET for rare diseases

→ 1 project selected for funding for a budget of **€ 2 million**

- **E-RARE 2** - transnational funding of collaborative, multidisciplinary projects on rare diseases - coordination  
(running until Sept 2014)

up to 2010

# Overview - Rare Diseases (FP7 - Area 2.4.4)



RareDiseasePlatform € 0.95 M

E-Rare-2 € 2 M



# Rare diseases: 2012 call for proposals

HEALTH-2012-2.4.4-1: Preclinical and clinical development of substances with clear potential as ODs

→ 10 projects selected for funding for a budget of ~ **€ 57 million**

- DEVELOPAKURE - Clinical Development of Nitisinone for **Alkaptonuria**
- PREVENTTORP – novel preventative intervention for the blinding disease **retinopathy of prematurity (ROP)**
- FIGHT-HLH - First Targeted Therapy to FIGHT **Hemophagocytic Lymphohistiocytosis (HLH)**
- MEUSIX - Clinical trial of gene therapy for **MPS VI**
- EUROFANCOLEN - Phase I/II Gene Therapy Trial of **Fanconi anemia** patients with lentiviral vector carrying the FANCA gene

# Rare diseases: 2012 call for proposals

## HEALTH-2012-2.4.4-2: **Observational studies**

→ 5 projects selected for funding for a budget of **15 € million**

- **BESTCILIA** - Better Experimental Screening and Treatment for **Primary Ciliary Dyskinesia**
- **OPTIMISTIC** - Observational Prolonged Trial In **Myotonic dystrophy** type 1 to Improve QoL-Standards
- **CHILD-EU** - Orphans Unite: chILD better together European Management Platform for **Childhood Interstitial Lung Diseases**
- **DESSCIPHER** - To decipher the optimal management of **systemic sclerosis**
- **DSD-LIFE** - Clinical European study on the outcome of surgical and hormonal therapy and psychological intervention in **disorders of sex development (DSD)**

# Rare diseases: 2012 call for proposals

HEALTH-2012-2.4.4-3: **Best practices and knowledge sharing in the clinical management of RDs**

→ 1 projects selected for funding – total budget **2 € million**

- RARE-BESTPRACTICE - Platform for sharing **best practices for clinical management** of RDs

# Rare diseases: 2012 call for proposals

## HEALTH-2012-2.1.1.1-A. Support for international rare diseases research

- SUPPORT IRDiRC - Support for international rare disease research to serve the IRDiRC objectives → **2 € million**

## HEALTH-2012-2.1.1.1-B. Clinical utility of -omics for better diagnosis of rare diseases

- NEUROMICS - Integrated European -omics research project for diagnosis & therapy in rare neuromuscular & neurodegenerat. diseases → **12 € million**
- EURENOMIS - European Consortium for High-Throughput Research in Rare Kidney Diseases → **12 € million**

# Rare diseases: 2012 call for proposals

## HEALTH-2012-2.1.1.1-C. Databases, biobanks and clinical 'bio-informatics' hub for rare diseases

- RD-CONNECT: An integrated platform connecting registries, biobanks and clinical bioinformatics for rare disease research → **12 € million**

# Rare diseases: 2013 call for proposals

HEALTH.2013.4.2-3: **New methodologies for clinical trials for small population groups**

➔ 2 projects selected for funding for a global budget of € **6 million**

- **IDEAL - Integrated DEsign and AnaLysis of small population group trials** (methods of general applicability irrespective of indication by Integrated DEsign and AnaLysis of clinical trials in SPG through a multidisciplinary closely collaborating consortium of researchers from European universities, research institutes and industry) - ***starting 1.11.2013***
- **ASTERIX - Advances in Small Trials dEsign for Regulatory Innovation and eXcellence** (validated innovative statistical design methodologies for cost efficient clinical trials deriving reliable results from trials in small population groups, especially focusing on rare diseases) – ***started 1.10.2013***

# Rare diseases: 2013 call for proposals

## HEALTH.2013.1.2-1: Development of imaging technologies for therapeutic interventions in rare diseases

→ 6 projects selected for funding for a total of € **34,5 million**

- BIOIMAGE-NMD - imaging technology to monitor response to novel therapies in **neuromuscular diseases (NMD)** using Duchenne muscular dystrophy (DMD) as exemplar disease
- COMMITMENT - Combined Molecular Microscopy for Therapy and Personalized Medication in **Rare Anaemias** Treatments
- MULTISYN - novel imaging system for combined simultaneous molecular and functional imaging (PET-MRI/fMRI) for **two rare diseases synusipathies**: multiple systems atrophy (MSA) and parkinsonism due to mutations in the alpha-synuclein gene (SNCA)

	<b>EXAMPLES of RD research from non RD-specific topics</b>	<b>n° projects</b>	<b>total budget (M€)</b>
<b>2011</b>	HEALTH-2011-1.4.1: <b>Regenerative medicine clinical trials</b>	1	5,2
	HEALTH-2011-1.4.2: <b>Tools, technologies and devices for application in regenerative medicine</b>	2	8,9
	HEALTH.2011.1.4-3: <b>Development and production of new, high-affinity protein scaffolds for therapeutic use</b>	1	5,9
	HEALTH-2011-2.1.1-4: <b>Population genetics studies in cardio metabolics disorders population in EU/AC and EECA</b>	1	3
	HEALTH-2011-2.2.1-1: <b>High impact initiative on human genome</b>	1	3
<b>2012</b>	HEALTH-2012-3.2-2: <b>New methodology for HTA</b>	1	3
<b>2013</b>	HEALTH.2013.0-1: <b>Boosting the translation of FP projects' results into innovative applications for health</b>	1	6
	HEALTH.2013.2.1.1-1: <b>Functional validation in animal and cellular models of genetic determinants of diseases and ageing processes</b>	1	12
	HEALTH.2013.1.4-1: <b>Controlling differentiation and proliferation in human stem cells intended for therapeutic use</b>	1	6



# Rare diseases: 2013 call for proposals – other areas (not RD-specific)

## HEALTH.2013.0-1: Boosting the translation of FP projects' results into innovative applications for health

- SCOPE-DMD - Consortium for Products across Europe in Duchenne Muscular Dystrophy → **6 € million**

## HEALTH.2013.2.1.1-1: Functional validation in animal and cellular models of genetic determinants of diseases and ageing processes

- SYBIL - Systems biology for the functional validation of genetic determinants of skeletal diseases → **12 € million**

## HEALTH.2013.1.4-1. Controlling differentiation and proliferation in human stem cells intended for therapeutic use

- REPAIR-HD - Human pluripotent stem cell differentiation, safety and preparation for therapeutic transplantation in Huntington's disease → **6 € million**

# Rare diseases: the trend in 2012 - 2013

- Until 2011 included **66 ongoing projects** worth **325 million €** EC funding
- In 2012 : **108 million €**
- In 2013 : not all project data has been disclosed
- However, from unofficial information, it appears that in the 2013 calls RD research was worth **above 100 million €** of which the majority (over 60 M€ ) under not specific RD topics
  
- **More than 200 million €** allocated to RD research in 2012 – 2013 by EC funds

# Rare diseases: 2013 call for proposals

## Conclusion:

- **Boosting of RD earmarked funds by EC in 2012 – 2013 : more than 200 € million**
- **Dramatic increase as from 2011 in both RD specific and not specific research (-omics and personalised medicine), where RD projects competed with non RD topics**
- **Response of DG Research, Health Directorate, to IRDiRC engagements**

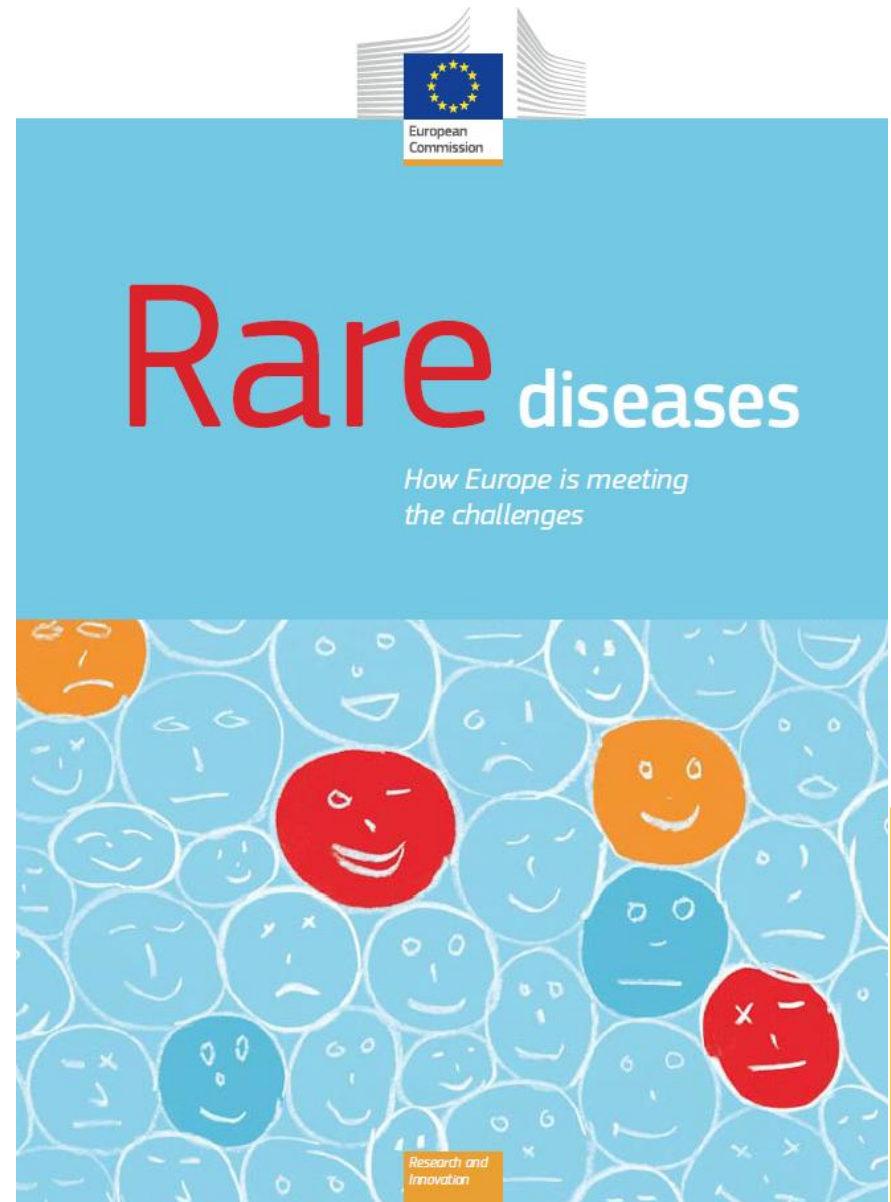
# Rare diseases in FP7

A selection of over 100 EU-funded projects from 2007 to 2013

(including DG Sanco programme on Public Health)

[http://ec.europa.eu/research/health/pdf/rare-diseases-how-europe-meeting-challenges\\_en.pdf](http://ec.europa.eu/research/health/pdf/rare-diseases-how-europe-meeting-challenges_en.pdf)

CORDIS (FP7 portal) « find a project »  
[http://cordis.europa.eu/fp7/projects\\_en.html](http://cordis.europa.eu/fp7/projects_en.html)



## 2. What do we know about **HORIZON 2020**

- New name for the **Framework Programme coming after FP7**, running from **2014 to 2020**
- Budget almost **70 billion**
- Brings together Framework Programme FP7, the Competitiveness and Innovation Programme (CIP) and EU's contribution to the European Institute of Technology (EIT)
  - Stronger focus on innovation and close-to-market activities
- **POLICY** : A core part of **EUROPE 2020**, Innovation and European Research Area:
  - Responding to the economic crisis
  - Addressing people's concerns
  - Strengthening EU global position
- **BUDGET** : Budget a core part of EU overall budget for 2014-2020

***EUROPE 2020** is the **EU's growth strategy** for this decade.*

*5 objectives by 2020: employment, innovation, education, social inclusion and climate/energy.*



Focus	Research	Research and Innovation
Budget	50 billion EUR	70,2 billion EUR
Programme	FP7 CIP EIT	HORIZON 2020
Pillars	Cooperation People Capacities Ideas	→ Excellent Science → Competitive Industries → Societal Challenges
In which pillar to find « health research »	<p style="text-align: center;"><b>“Health”</b> part of the Cooperation programme</p>	<p style="text-align: center;"><b>“Health, demographic &amp; wellbeing challenge”</b> part of “Societal Challenges “</p>
	<p style="text-align: center;">European Research Council part of “Ideas”</p>	<ul style="list-style-type: none"> <li>• European Research Council</li> <li>• Marie Skłodowska Curie Actions</li> <li>• Research Infrastructures</li> </ul> <p style="text-align: center;">part of “Excellent Science”</p>
	<p style="text-align: center;">Marie Curie Actions part of “People”</p>	
	<p style="text-align: center;">Research Infrastructures part of “Capacities”</p>	

22 300 M€

## → Excellent Science

Aim:  
Science base

1. European Research Council **11 900 M€**
2. Future & Emerging Technologies (FET)
3. Marie Skłodowska Curie Actions
4. Research infrastructures **5 600 M€**

2 300 M€

Joint  
Research  
Centre

15 500 M€

## → Competitive Industries

Aim:  
Growth

1. Key Enabling Technologies (KET) **12 400 M€**
2. Access to Risk Finance
3. Innovation in SMEs

European Institute  
of Technology

27 000 M€

## → Societal Challenges

Aim:  
Citizens' concerns

1. Health **7 400 M€**
2. Bio-economy
3. Energy
4. Transport
5. Climate action
6. Integrative, innovative societies
7. Secure societies

IMI

SCIENCE for and with SOCIETY





→ Excellent Science

24 598 M€

WHY? to attract and retain research talent and provide access to researchers to best infrastructures

**1. European Research Council**

- Frontier research for best individual teams

13 000 M€

**2. Marie Skłodowska Curie Actions (MSCA)**

- Opportunities for training and career development

5 700 M€

**3. Research infrastructures**

- Securing access to world-class facilities

2 500 M€

## → Competitive Industries

17 938 M€

WHY? to underpin innovation across existing and merging sectors, attract more private investments and support innovative SMEs

### 1. Key Enabling Technologies

- ICT, nanotechnologies, **biotechnology**, materials, etc.

13 781 M€

### 2. Access to risk finance

- Leveraging private finance and venture capital for research and innovation

3 500 M€

### 3. Innovation in SMEs

- Fostering innovation in all types of SMEs
- (extra budget 15% from Societal Challenges)

0,6 M€

## → Societal Challenges

27 000 M€

WHY? To face concerns of citizens and society and implement EU policy objectives that need innovation.

HOW?

- multidisciplinary collaborations including social science and humanities
- Promising solutions to be tested demonstrated and scaled up

### 1. Health

- Brings together FP7 Health and ICT in Health
- Builds on eHealth, ICT & Aging, AAL (ambient assisted living)

7 400 M€

2. Bio-economy
3. Energy
4. Transport
5. Climate action
6. Integrative, innovative societies
7. Secure societies

## **CROSS-DISEASE THEMES !**

- Understanding health, wellbeing and disease
- Preventing disease
- Treating and managing disease
- Active aging and self management of health
- Methods and data
- Health care provision and integrated care

→ **Change in Horizon 2020 – Health Programme :**

- **NO LONGER Disease Specific topics = NO LONGER RARE DISEASE specific topics (with exceptions)**
- **Cross-disease THEMES = RD specific project competing with all other diseases**

→ In practice :

- **HEALTH Specific Programme under the pillar “Societal Challenges”**

→ Each Specific Programme will rely on

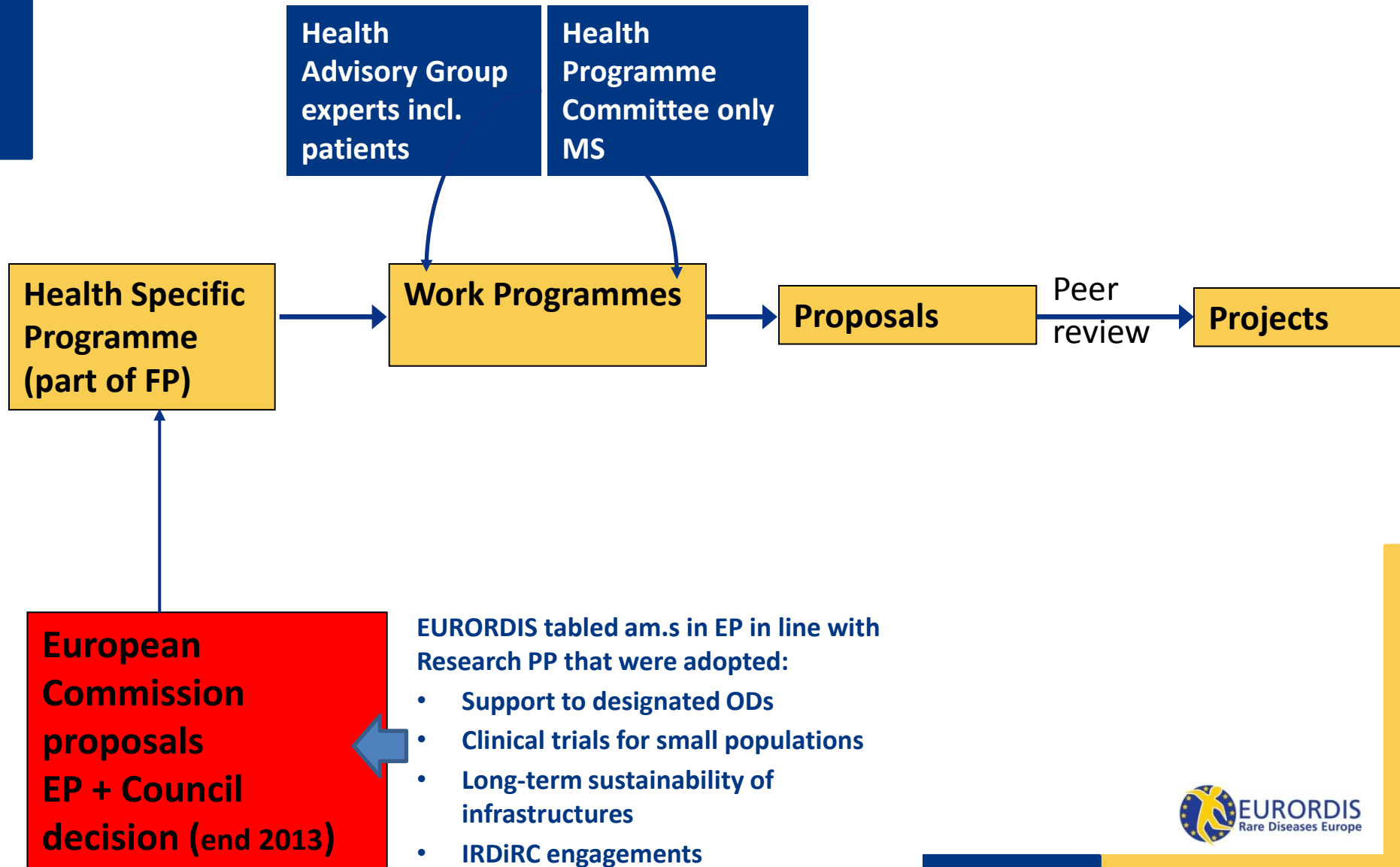
- **Strategic Programmes** (3-year duration) and
- **Work Programmes** (2-year duration)

# Proposed Work Programme Cycle

2014	2015	2016	2017	2018	2019	2020
Strategic Programme						
Work Programme 1 (plus tentative information for 2016)		Strategic Programme				
		Work Programme 2 (plus tentative information for 2018)		Strategic Programme		
				Work Programme 3 (plus tentative information for 2020)		
						Work Programme 4

**HEALTH Specific Programme**

# FROM THE PROGRAMME TO THE PROJECTS



EURORDIS tabled am.s in EP in line with Research PP that were adopted:

- Support to designated ODs
- Clinical trials for small populations
- Long-term sustainability of infrastructures
- IRDiRC engagements

# HEALTH ADVISORY GROUP

Name: Horizon 2020 Advisory group for Health, demographic, change and wellbeing (E02942)

## Individual expert appointed in his/her personal capacity

Name	Nationality	Professional Title
<a href="#">Adrian Saftoiu</a>	Romania	
<a href="#">Anders Olauson</a>	Sweden	
<a href="#">Andriana Prentza</a>	Greece	
<a href="#">David Norris</a>	United Kingdom	
<a href="#">Elias Mossialos</a>	Greece	
<a href="#">Elisabete Weiderpass</a>	Sweden	
<a href="#">Ellen Fritsche</a>	Germany	
<a href="#">Geneviève Inchauspe</a>	France	
<a href="#">Georges De Moor</a>	Belgium	
<a href="#">Hele Everaus</a>	Estonia	
<a href="#">Jackie Hunter</a>	United Kingdom	
<a href="#">Leszek Kaczmarek</a>	Poland	
<a href="#">Lora Fleming</a>	United States of America	
<a href="#">Lucia Monaco</a>	Italy	
<a href="#">Marie-Paule Kiény</a>	France	
<a href="#">Marta Cascante</a>	Spain	
<a href="#">Martine Piccard</a>	Belgium	
<a href="#">Matej Oresic</a>	Slovenia	
<a href="#">Mathieu Simon</a>	France	
<a href="#">Orla Sheils</a>	Ireland	
<a href="#">Paolo Lisboa</a>	Portugal	
<a href="#">Paula Marques Alves</a>	Portugal	
<a href="#">Peter Saraqa</a>	United Kingdom	
<a href="#">Petra Wilson</a>	United Kingdom	
<a href="#">Rafael Benqoa</a>	Spain	

→ **Health Advisory Group in the Transparency Registry of the EC**

<http://ec.europa.eu/transparency/registries/expert/index.cfm?do=groupDetail.groupDetail&groupID=2942>

- **Independent experts** consulted to prepare Work Programmes
- Scientific and industry experts in their own capacity

## - Organisation

Name of Organisation	Category	Countries / Areas represented
<a href="#">School of Public health, University of Debrecen</a>	Academia	Hungary
<a href="#">Swiss Tropical Institute, University of Basel</a>	Academia	Switzerland
<a href="#">The BioRobotics Institute</a>	Academia	Italy
<a href="#">TILDA - Trinity College Dublin</a>	Academia	Ireland
<a href="#">University Copenhagen</a>	Academia	Denmark
<a href="#">University of Oslo</a>	Academia	Norway



## → Health Programme Committee ???

- **No members' name and organisations published in the Transparency Registry of the EC !**

- **Member States' forum** to give input to Commission on Work Programmes.
- Approve the **final version of the Work Programmes** before calls are published
- Usually research agencies' representatives, e.g. MRC, INSERM etc.
- Link between the EU and national research systems

→ **HORIZON 2020**

- July 2013 : Overall EU budget, informal agreement 2014-2020 (EP & Council)
- November 2013 : EP and Council formally adopt EU overall budget
- November/December 2013: EP and Council formally adopt HORIZON 2020
- **11 December 2013**: adoption of WP and publication of the calls for proposals

**→ WORK PROGRAMME**

- January 2013: First Strategic Programme
- February 2013: First meetings of the Horizon 2020 Advisory Groups
- July 2013: Consultation of with Health Programme Committees (MS)
- Summer 2013: Draft first Work Programme 2014-2015
- Autumn 2013: EC inter-service consultation and consultation of the Health Programme Committee (MS)
- **11 December 2013**: Adoption of the Work Programme and publication of the calls for proposals

## Unofficial information ! Possible topics:

- Health promotion and disease prevention: translating -omics into stratified approaches
- Evaluating screening and prevention programmes
- New diagnostics tools and technologies: in vitro devices, assays and platforms (2014) & in vivo medical imaging technologies (2015)
- Clinical validation of biomarkers
- **New therapies for RD (possibly 2015)**
- Tools and technologies for advanced therapies
- Effectiveness of healthcare interventions in pediatric population
- Pilots on personalised medicine in health and care systems
- Self-management of health and disease: citizen engagement and patient empowerment supported by ICT
- Support to ERNs efficient network modelling & validation
- **ERA-NET RD research implementing IRDiRC objectives**

Exception to general rule  
of cross-cutting topics !  
IRDiRC engagements

## How each topic will look like:

- Simplified, less detailed topic description to allow greater freedom to applicants. Should allow innovative approaches to broaden participation
- Structure of each topic:
  - Specific challenge
  - Scope
  - Extended impact
- Type of action: usually Collaborative Project or Support Action (100% funding)
- Topics divided in the main areas of Health SP
- **Focus on personalised health and care, for sustainability and equity in European health and care systems**
- Budget roughly 1 billion EUR



<b>Funding rates</b>	<b>Research activities MAX 75%</b>	<b>Research activities MAX 100%</b> (except for Innovation actions, where a 70% maximum will apply for profit making entities)
	<b>Overhead 20%, 60% or actual</b>	<b>Overhead 25%</b>
<b>Time to grant</b>	<b>12 months average</b> from submission of proposal	<b>8 months average</b> from submission of proposal

- Moving from several funding rates for different beneficiaries and activities to just two
- **Single flat rate** to calculate **overheads** or «indirect costs»
- Successful applicants to get working more quickly: reduction of average time to grant by 100 days

→ In practice :

- **Grants for RESEARCH and INNOVATION**

→ Funding of all activities and participants: 100%

→ Minimum 3 participants

→ Main type of activity: Research and Development

→ Project types:

- **Collaborative projects**, from large integrated projects to small targeted research projects

→ In practice :

- **SUPPORT and COORDINATION ACTION**

→ Funding of all activities and participants: 100%

→ Minimum 1 participant

→ Main type of activity: Support and coordination activities, e.g. accompanying measures, studies, networking, etc.

→ Project types:

- **Coordination and Support Actions**



→ In practice :

- **ETHICS & OPEN ACCESS**

→ Ethics rules remain unchanged from FP7, notably rules on embryonic stem cells.

- No funding is granted for research activities in a country where such activity is forbidden
- All research must comply with ethical principles and relevant national (& EU & international legislation)

→ Stronger emphasis on open access of publication funded by H2020 (to avoid duplication and optimising investment)

- Open access mandatory for all publications resulting from EU-funded research
- Costs of publications considered as eligible costs

→ In practice :

- **INTERNATIONAL COOPERATION**

→ Crucial to address many Horizon 2020 objectives (e.g. IRDiRC)

→ Principle of general openness: Cross-cutting theme across H2020

→ Horizon 2020 shall be open to the association of:

- acceding countries, candidate countries and potential candidates
- selected international partner countries that fulfill the relevant criteria (capacity, track record, close economic and geographical links to the Union, etc.)

1. International collaboration on RD research: **IRDIRC**, an *ad hoc* consortium

[http://ec.europa.eu/research/health/medical-research/rare-diseases/irdirc\\_en.html#anch\\_vid](http://ec.europa.eu/research/health/medical-research/rare-diseases/irdirc_en.html#anch_vid)

## → Why an international consortium ?

**Research challenges related to rare diseases are too big to be mastered alone**

- Huge unmet medical needs for patients across the globe
- Small patient populations
- Scarce and scattered research resources and expertise, etc.

## → Why working together ?

- Mobilise the necessary critical mass of expertise and resources
- Avoid overlaps in research allowing for more diseases to be tackled
- Deliver new cures and diagnoses to treat patients world-wide

*from the Welcome Address of  
Ruxandra Draghia Akli , Director of DG Research  
for Health, at the IRDiRC first Conference  
Dublin, April 2013*

→ IRDiRC vision

2020 goals in RD research

→ 200 new therapies

→ Means to diagnose most RDs

## NEW THERAPIES



Objective 2020: 200 new therapies

Disclaimer: the numbers do not reflect IRDiRC initiatives only

[www.irdirc.org](http://www.irdirc.org)

## 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)**

**EURORDIS**



## Asia

**BGI (China)**

**Chinese RD Research  
Consortium (China)**

## Australia

**Western Australian  
Department of Health**

## 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)**

**NORD**

**Genetic Alliance**

# IRDIRC GOVERNANCE

## Patient representatives

**Executive Committee**

← **Béatrice de Montleau**  
**Marie C. Ouillade** (AFM)  
**Peter Saltonstall** (NORD)  
**Sharon Terry** (GA)



## Scientific Committees

**Diagnostics**  
Chair **Kym BOYCOTT**

**Interdisciplinary**  
Chair **H. Lochmüller**

**Therapies**  
Chair ...

← **Maria Mavris**  
**Alastair Kent**  
(GA)



## Working Groups

**Genome/Phenome**

**Data sharing/  
Bioinformatics**

**Regulatory**

← **Yann Le Cam**

**Model systems**

**Biobanks**

**Repurposing**

← **Nick Sireau**  
(DevelopAKUre)

**Ontologies**

**Ethics and  
governance**

**Cell and Gene  
Therapies**

← **Tsveta Schyns**  
(nEUROPED)

**Sequencing**

**Registries and  
natural history**

**Biomarkers**

← **Anna Kole**  
(RD-Connect)

**SUPPORT IRDiRC Secretariat – EU funded project -FP7**  
**Coordinator Prof. Ségolène Aymé**



## 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**



- **Governance** just reviewed
- **Working on a Policy Paper** - a framework addressing various aspects of research:
  - Sharing data and samples
  - Common quality standards
  - Interoperability and harmonisation of ontologies and biobanks
  - Rapid release of research results, etc.
- **Defining GAP ANALYSIS** – where international research should focus its attention
  - Defining and counting what members fund or have funded
  - Then identifying gaps

# What IRDiRC members fund e.g.

- **Genome Canada:** IGNITE project, worth 5 million Can \$, as well as Care 4 Rare & FORGE with the CIHR (Canadian Institutes for Health Research) on gene identification;
- **NIH:** National Eye Institute: 75 million \$ in RD related projects (a programme has just been launched on rare eye diseases and molecular therapies for eye diseases);
- **FDA** - Office for Orphan Product Development: budget for grants worth 14 million \$/year
- **Instituto de Salud Carlos III**, Spain: approx. 50 projects funded in the field of RDs
- **Telethon Italia** – bottom-up calls worth 10.5 MEUR/year, with 38 new grants in the last call. Plus a new type of call for “exploratory projects” (diseases that are not addressed otherwise, inputs from POs)
- **European Commission:** 530 million EUR and 103 projects funded under FP7 (7 years)

# IRDIRC TIMELINE



**Launch of  
IRDIRC 2011**

**IRDIRC  
Conference**

**Scientific  
Committees**



**3000 diagnostics**



**6000 diagnostics**

**YEAR**

**2012**

**Working  
Groups**

**2015**

**50 new applications for  
market authorisation**

**2020**

**200 new applications for  
market authorisation**



# WRAP-UP

- RD research funding in FP7 doubled: 530 M€, with a significant increase in the last 2-3 years
- Health Specific Programme in H2020 approx. 7000 M€
- 100% funding rate all beneficiaries all activities, 25% flat rate for overheads
- RD remain a priority in Health Specific Programme, in line with IRDiRC engagements
- However no RD-specific topics, only cross-cutting topics (with some exceptions)
- WP 2014-15 calls out on 11<sup>th</sup> December 2013
- Next WP: think of contacting your (national) representative at the Health Programme Committee or Health Advisory Group

# NATIONAL INFORMATION EVENTS

Austria	21 January 2014	<a href="http://www.era.gv.at">www.era.gv.at</a>
Belgium	15 October 2013	<a href="http://www.ncpwallonie.be">www.ncpwallonie.be</a>
Belgium	14 November 2013	<a href="http://www.h2020-kickoff-belgium.be/">www.h2020-kickoff-belgium.be/</a>
Bulgaria	17 February 2014	coming soon
Croatia	10 February 2014	coming soon
Cyprus	16 January 2014	coming soon
Czech Republic	18 October 2013	<a href="http://www.fp7.cz/cs/conference-teera">www.fp7.cz/cs/conference-teera</a>
Denmark	29 October 2013	<a href="http://fivu.dk">http://fivu.dk</a>
Estonia	31 January 2014	<a href="http://www.etag.ee">www.etag.ee</a>
Finland	11 October 2013	<a href="http://www.tekes.eu">www.tekes.eu</a>
France	16 December 2013	<a href="http://www.horizon2020.gouv.fr">www.horizon2020.gouv.fr</a>
Germany	28-29 January 2014	<a href="http://www.forschungsrahmenprogramm">www.forschungsrahmenprogramm</a>
Greece	10 January 2014	coming soon
Hungary	26-27 November 2013	<a href="http://www.nih.gov.hu/">www.nih.gov.hu/</a>
Ireland	10 December 2013	coming soon
Italy	27 February 2014	coming soon
Latvia	24 January 2014	coming soon
Lithuania	4 December 2013	<a href="http://www.vif2013.com">www.vif2013.com</a>
Luxembourg	14 January 2014	<a href="http://www.innovation.public.lu">www.innovation.public.lu</a>
Netherlands	12 November 2013	<a href="http://www.agentschapnl.nl">www.agentschapnl.nl</a>
Norway	6 December 2013	coming soon
Poland	7 November 2013	coming soon
Portugal	13 December 2013	coming soon
Romania	3-4 October 2013	<a href="http://Conferinta.nationala">Conferinta nationala</a>
Slovakia	21 January 2014	coming soon
Slovenia	29 November 2013	coming soon
Spain	11-12 November 2013	<a href="http://Centro de Congresos Príncipe Felipe">Centro de Congresos Príncipe Felipe</a>
Sweden	29 January 2014	coming soon
Switzerland	14-17 January 2014	<a href="http://www.launch-h2020.ch/">www.launch-h2020.ch/</a>
United Kingdom	31 January 2014	coming soon

[http://ec.europa.eu/research/horizon2020/index\\_en.cfm?pg=h2020-events](http://ec.europa.eu/research/horizon2020/index_en.cfm?pg=h2020-events)

**EU-wide in Brussels:  
22 November 2013**



- **HORIZON 2020**

[http://ec.europa.eu/research/horizon2020/index\\_en.cfm?pg=home](http://ec.europa.eu/research/horizon2020/index_en.cfm?pg=home)

EU research

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

- **EU Health Research**

[http://ec.europa.eu/research/health/index\\_en.html](http://ec.europa.eu/research/health/index_en.html)

- **EU Rare Disease Research**

[http://ec.europa.eu/research/health/medical-research/rare-diseases/index\\_en.html](http://ec.europa.eu/research/health/medical-research/rare-diseases/index_en.html)

- **IRDiRC**

[http://ec.europa.eu/research/health/medical-research/rare-diseases/irdirc\\_en.html](http://ec.europa.eu/research/health/medical-research/rare-diseases/irdirc_en.html)

[www.irdirc.org](http://www.irdirc.org)

- **Information on research programmes and projects**

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

THANK YOU

QUESTIONS & ANSWERS

