EU Research Programmes
Funding rare disease research

Irene Norstedt, Head of Innovative and personalised Medicine Unit, Research & Innovation DG, European Commission

ECRD, 9 May 2014
Rare diseases

How Europe is meeting the challenges
The EU: A major player in funding collaborations for rare diseases research

Over two decades of investment in the area

7th EU Research Framework Programme (2007-2013):

- Over €620 million invested in close to 120 collaborative projects
- Plus more than 100 individual fellowships, grants and training networks

47 projects
€ 64 million

59 projects
€ 230 million

~120 projects
>€ 620 million
EU funded collaborative research in rare diseases

- Europe wide studies of natural history and pathophysiology
- In vitro/in vivo models
- Registries & bio-banks
- Identification of biomarkers
- Clinical trials methodologies for small populations
- -omics for rare diseases and linking data
- Development of preventive, diagnostic and therapeutic interventions
2012-2013 FP7 Health Calls

8 research topics for rare diseases:

- Omics for better understanding of diseases in view of development of new diagnostics and treatments *
- Databases, biobanks and clinical ‘bio-informatics’ hub
- Preclinical and clinical development of orphan drugs *
- Observational trials in rare diseases
- Best practice and knowledge sharing in clinical management
- Organisational support for IRDiRC
- Development of imaging technologies for therapeutic interventions in rare diseases *
- New methodologies for clinical trials for small population group

*industry/SME topic
New Orphan Drugs

• 17 projects with an EU investment of € 80 million
• Several disease areas including neurology, immunology, and dermatology
• Call topic focus:
  • Preclinical and/or clinical development of substances with a clear potential as orphan drugs
  • Diagnostics and therapies for rare diseases
## New Orphan Drugs

<table>
<thead>
<tr>
<th>Project</th>
<th>Disease</th>
<th>Therapy</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>MABSOT</strong></td>
<td>Delayed graft function</td>
<td>Antibody OPN305 - OMP designation for use in the prevention of DGF</td>
</tr>
<tr>
<td><strong>IMPACTTT</strong></td>
<td>Cystic Fibrosis</td>
<td>Anti-pseudomonas IgY</td>
</tr>
<tr>
<td><strong>AIPgene</strong></td>
<td>Acute intermittent porphyria</td>
<td>Gene therapy using AAV5-AAT-PBGD to deliver porphobilinogendeaminase (PBGD) expression cassette directly into hepatocytes</td>
</tr>
<tr>
<td><strong>GENE-GRAFT</strong></td>
<td>Recessive dystrophic epidermolysis bullosa</td>
<td>Skin equivalent graft genetically corrected with a COL7A1-encoding SIN retroviral vector</td>
</tr>
</tbody>
</table>
# New Orphan Drugs

<table>
<thead>
<tr>
<th>Project</th>
<th>Disease</th>
<th>Therapy</th>
</tr>
</thead>
<tbody>
<tr>
<td>FIGHT-HLH</td>
<td>Hemophagocytic lymphohistiocytosis</td>
<td>Fully humanized anti-IFNγ mAb</td>
</tr>
<tr>
<td>PROFNAIT</td>
<td>Fetal/Neonatal Alloimmune Thrombocytopenia</td>
<td>Anti Human Platelet Antigen-1a immunoglobulin (IgG) (Tromplate®)</td>
</tr>
<tr>
<td>PREVENTROP</td>
<td>Blinding disease retinopathy of prematurity</td>
<td>The growth factor complex IGF-I/IGFBP-3 (Premiplex®)</td>
</tr>
<tr>
<td>EURO-FANCOLEN</td>
<td>Fanconi anemia</td>
<td>Genetic correction of hematopoietic stem cells (HSC) utilising novel lentiviral vector FANCA-LV</td>
</tr>
</tbody>
</table>
## New Orphan Drugs

<table>
<thead>
<tr>
<th>Project</th>
<th>Disease</th>
<th>Therapy</th>
</tr>
</thead>
<tbody>
<tr>
<td>Develop-AKUre</td>
<td>Alkaptonuria</td>
<td>Assessing nitisinone as potential treatment</td>
</tr>
<tr>
<td>Traumakine</td>
<td>Acute lung injury</td>
<td>Biopharmaceutical interferon-beta (IFN-beta) (EU3/07/505)</td>
</tr>
<tr>
<td>Net4CGD</td>
<td>X-linked chronic granulomatous disease</td>
<td>Autologous haematopoietic cells genetically modified with a lentiviral vector containing human gp91 gene</td>
</tr>
<tr>
<td>MeuSIX</td>
<td>Mucopolysaccharidosis VI</td>
<td>AAV vector-mediated gene therapy expressing human ARSB gene</td>
</tr>
<tr>
<td>STRONG</td>
<td>Neovascular glaucoma</td>
<td>Topical treatment of Aganirsen antisense oligonucleotide</td>
</tr>
</tbody>
</table>
## New Orphan Drugs

<table>
<thead>
<tr>
<th>Project</th>
<th>Disease</th>
<th>Therapy</th>
</tr>
</thead>
<tbody>
<tr>
<td>ODAK</td>
<td>Acanthamoeba keratitis</td>
<td>Preclinical and clinical research of Polihexanide</td>
</tr>
<tr>
<td>MYASTERIX</td>
<td>Myasthenia gravis</td>
<td>Therapeutic vaccine candidate</td>
</tr>
<tr>
<td>PADDINGTON</td>
<td>Huntington’s Disease</td>
<td>Disease-modifying properties of SEN0014196 (novel selective SirT1 inhibitor)</td>
</tr>
<tr>
<td>RDCVF</td>
<td>Retinitis pigmentosa</td>
<td>Rod-derived Cone Viability Factor</td>
</tr>
<tr>
<td>PRATH</td>
<td>Atypical Haemolytic Uraemic Syndrome</td>
<td>Recombinant human minibody against Complement component C5</td>
</tr>
</tbody>
</table>
Clinical trials for rare diseases

- Innovative statistical design methodologies for clinical trials in small populations focussing on rare diseases
- 3 projects bringing together international experts in innovative clinical trial design methodology along with key stakeholders
IDEAL: Integrated DEsign and AnaLysis of small population group trials

- Assessment of randomisation procedures
- Extrapolating dose-response information
- Optimal designs in mixed models
- Pharmacogenetic designs
- Simulation of clinical trials
- Decision analysis and biomarker surrogate endpoints

Coordinator: Ralf-Dieter Hilgers, Aachen University
InSPiRe: Innovative methodology for small populations research

- Early phase dose-finding studies
- Decision-theoretic methods for clinical trials
- Use of evidence synthesis in the planning and interpretation of clinical trials

Coordinator: Nigel Stallard, Warwick Medical School
ASTERIX: Advances in Small Trials Design for Regulatory Innovation and Excellence

- New standards of evidence that take into account the rare prevalence of disease
- Enabling adaptive designs and sequential meta-analysis using multiple endpoints
- Providing a blueprint to pro-actively share information on trials in the planning stage

Coordinator: Kit Roes, Utrecht University
"-Omics" for rare diseases

- 2 projects focusing on molecular characterisation using -omics technologies.
- Ontologies, reference -omics profiles, diseases models, development of technologies
- New means to diagnose and allow development of new treatments for these diseases
- Platform for integrating -omics data with clinical data, connecting registries, biobanks and clinical bioinformatics
- Provides access to -omics profiles and samples
• The EU’s 2014-20 programme for research & innovation (around € 80 billion)
• A core part of Europe 2020, Innovation Union & European Research Area
• Three priorities: Excellent science, Industrial leadership, Societal challenges
Horizon 2020 - what's new?

• A single programme bringing together three separate programmes/initiatives*

• Coupling research to innovation – from research to retail, all forms of innovation

• Focus on societal challenges facing European society e.g. health and ageing, clean energy and transport

• Simplified access, for all companies, universities, institutes in all EU countries and beyond

*The 7th Research Framework Programme (FP7), innovation aspects of Competitiveness and Innovation Framework Programme (CIP), EU contribution to the European Institute of Innovation and Technology (EIT)
Getting you started faster

• A single set of simpler and more coherent participation rules

• Just 2 funding rates (70% or 100%)

• Single flat rate (25%) for overhead or 'indirect costs'

• Simpler financial regulation to come

• 8 months time-to-grant target
Health, demographic change and wellbeing challenge

- Translate science to benefit citizens
- Test and demonstrate new healthcare models, approaches and tools
- Promote healthy and active ageing
- Improve health outcomes, reduce inequalities
- Support a competitive health sector

Over €7 billion to health research
A new approach

- Challenge-driven
- Broader topics
- Two-year work programme
- Stronger focus on end users
- eHealth fully integrated
Focus areas of 2014-2015 Work Programme

• Understanding health, ageing & disease
• Effective health promotion, disease prevention, preparedness and screening
• Improving diagnosis
• Innovative treatments and technologies
• Advancing active and healthy ageing
• Integrated, sustainable, citizen-centred care
• Improving health information, data exploitation and providing an evidence base for health policies and regulation
Topics for Rare disease research in 2014-2015 Work Programme

ERA-NET: Rare disease research implementing IRDiRC objectives (WP 2014)

New therapies for rare diseases (WP 2015)

Support for European Reference Networks: efficient network modelling and validation (WP 2015)

Clinical validation of Biomarkers (SME instrument: open call)
Other topics relevant to rare diseases

- Understanding health .. and disease ..
- Understanding diseases: systems medicine
- ..disease prevention .. translating 'omics' ..
- ..evaluating screening programmes ...
- ..new diagnostic tools .. in vitro ..
- ..tools ..technologies ..advanced therapies
- Clinical research ... regenerative medicine

See also other Work Programmes

Infrastructures ('starting communities' grant)
Marie Skłodowska-Curie Actions
European Research Council
Rare Disease Research in Horizon 2020

- Continued strong support and commitment to meet IRDiRC goals
- Focus on bringing novel therapies and diagnostics to patients
- Integrated approach with several funding opportunities/instruments
Innovative Medicines Initiative 2

- 3.45 billion EUR, equally shared between public and private partners
- For breakthrough vaccines, medicines and treatments
- For top quality research and innovation with great public health benefits and commercial possibilities
Active and Assisted Living 2

- Market-oriented ICT research in assisted living technologies and services
- New instruments e.g. prizes and innovation grants
- More EU funding of up to €175 million
- Participating countries pay ~€225 million
Research and Innovation

Participant Portal – your one-stop shop

RESEARCH & INNOVATION

Participant Portal

European Commission > Research & Innovation > Participant Portal > Home

HOME FUNDING OPPORTUNITIES HOW TO PARTICIPATE EXPERTS SUPPORT

Participant Portal

European Commission

Horizon 2020 Funding
Starting from 1/1/2014

On this site you can find and secure funding for research & innovation projects under the following EU programmes:

- 2014-2020 Horizon 2020 - research and innovation framework programme
- 2007-2013 7th research framework programme (FP7) and Competitiveness & Innovation Programme (CIP)

Non-registered users

- search for funding
- read the H2020 Online Manual & download the legal documents
- check if an organisation is already registered
- contact our support services or check our FAQs

Registered users

- submit your proposal
- sign the grant
- manage your project throughout its lifecycle

http://ec.europa.eu/research/participants/portal
Thank you

Irene.norstedt@ec.europa.eu

http://ec.europa.eu/research/health
http://ec.europa.eu/programmes/horizon2020