Research on Rare Diseases: DG RTD Initiatives

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European Commission
Research Directorate General
Health Directorate - Major Diseases Unit
1. From the 5th to the 6th Framework Programme

2. Rare Diseases: present projects and future prospects
1. From the 5th to the 6th Framework Programme

- Strengthen S&T bases of industry + encourage international competitiveness
- Promote research activities in support of other EU activities

Treaty establishing the EC → FP
Rare diseases in FP5

Specific programme ‘Improving the Quality of Life and Management of living resources (Quality of Life)’

a. Key actions (Food, nutrition and health; Control of infectious diseases; The "cell factory"; Environment and health; Sustainable agriculture, fisheries and forestry and integrated development of rural areas including mountain areas; The ageing population and disabilities)

b. Research and technological development activities of a generic nature
- Chronic and Degenerative Diseases, Cancer, Diabetes, Cardiovascular Diseases and Rare Diseases
- Research into Genomes and Diseases of Genetic Origin

c. Support for research infrastructures
5th -> 6th Framework Programme

- GA7
- GA8
- INFRA

34 projects, € 45 million
Previous programmes:
Promote cooperation and collaboration

FP6:
Develop scientific and technical excellence

↓
Make a reality of the European Research Area, with a view to stepping up innovation in Europe, in conjunction with all the efforts made to this end at national, regional and European level.
## Integrating European Research

### Thematic Priorities

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### Specific Activities Covering a Wider Field of Research

- Supporting EU policies
- Anticipating scientific and technological needs
- Horizontal research activities involving SMEs
- Specific measures in support of international cooperation
- Non-nuclear activities of the Joint Research Centre

## Structuring the European Research Area (ERA)

| Research & Innovation | Human res., mobility | Research infrastruct. | Science and Society |

## Strengthening the Foundations of ERA

| Coordination of activities | Development of research/innovation policies |
1.1 Advanced genomics and its application for health (€ 1100 M)

1.1.1 **Fundamental** knowledge and basic tools for functional genomics in all organisms

1.1.2 **Applications** of knowledge and technologies in the field of genomics and biotechnology for health

1.2 Combating major diseases (€ 1155 M)

1.2.1 Application-oriented genomic approaches to medical knowledge and technologies

1.2.2 Combating **cancer** (400 M)

1.2.3 Confronting the **major communicable diseases** linked to poverty

755 M

400 M
What is new compared to FP5?

- Increase in the global budget (€ 17.5 billion)
- Focus on thematic priorities
- New instruments
- More flexibility and autonomy
- Participation + funding of Third Countries
Proposal submission

Eligibility check

Individual evaluation/review

Eligible

Consensus meeting(s)

ESR

below a threshold

Panel evaluation

ESR

CR

above thresholds

Priority list

Commission decision

Ethical review

Commission decision

Report

Rejection

Reserve list

Negotiation
2. Rare Diseases: present projects and future prospects

a. Some FP5 projects

b. Topics open for funding in the two first calls of FP6
Friedreich's Ataxia (QLG1-1999-00584)
Molecular and Biochemical Pathogenesis of Friedreich's Ataxia: Search for Treatments

• RTD project
• 3 years
• 5 participants (French Co-ordinator)
• Aim: identify treatments and outcome measures for multicenter treatment trials for Friedreich’s ataxia, by developing mouse and cell culture models of FRDA, which will be used to identify mechanisms by which reduced expression of frataxin, or expression of mutated frataxin causes cell death.
MitEURO (QLG1-2001-00966)
Concerted Action on Mitochondrial Biogenesis and Disease

- Concerted action
- 3 years
- 39 participants (Finnish Co-ordinator)
- Aim: characterisation of the components of the apparatus of mitochondrial biogenesis in key model organisms, to establish and exploit pathogenic models in cell culture and in whole organisms, and to develop techniques for manipulating mitochondrial gene expression, aimed at eventual therapy. The consortium is pooling research tools and resources, promoting researcher exchanges and training, and enhancing scientific communication between scientists with complementary expertise.
CF-PRONET (QLG1-2001-01005)
Cystic fibrosis: rescue of the function and of the processing of CFTR mutants by pharmacological agents and by interacting proteins

• RTD project
• 3 years
• 9 participants (Belgian Co-ordinator)
• Aim: design compounds capable of correcting the processing and dysfunction of CFTR, characterise known interacting proteins and novel interacting proteins, at the genetic, biochemical and electrophysiological level and determine their ability to compensate for or rescue the cells from dysfunction.
Regiscar (QLG1-2002-01738) European registry of severe cutaneous adverse reactions (SCAR) to drugs and collection of biological samples

- RTD project
- 3 years
- 14 participants (German Co-ordinator)
- Aim: develop a European registry of SCAR (to allow pharmacoepidemiology and continuous surveillance of new drugs), a centralised collection of biological samples (to investigate the immunologic mechanisms of SCAR, and to look for candidate genes), and a large cohort of patients (to provide insights on the outcome, impact of sequelae on the quality of life and possible treatments of these reactions).
Eurobiobank (QLR1-2002-02769)  
European network of DNA, cell and tissue banks on rare diseases

• Thematic network
• 3 years
• 16 participants (French Co-ordinator)
• Aim: optimise the exploitation of biological collections of human material, and help reaching a critical mass of collections indispensable for research on rare diseases. Activities will focus on: development of a dedicated website interconnecting and facilitating access to collections; definition of high quality criteria and Standard Operating Procedures; mapping of relevant practical ethical issues; dissemination of SOP and know-how to the whole European scientific community.
# Integrating European Research

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## Strengthening the Foundations of ERA

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The strategic objective of this line is to develop improved strategies for the prevention and management — using also advanced technologies for health — of human disease and for living and ageing healthily. It will concentrate exclusively on integrating genomic approaches through all relevant organisms into more established medical approaches for investigating disease and health determinants. The emphasis will be put on translational research aimed at bringing basic knowledge through to clinical application.

*Council Decision 30/09/02*
Combating cardiovascular disease, diabetes, and rare diseases:

The objectives are to improve the prevention and management of important causes of mortality and ill health in Europe and to pool Europe's research resources for tackling rare diseases. Research will focus on: integrating clinical expertise and resources with relevant model systems and advanced tools in functional genomics to generate breakthroughs in the prevention and management of these diseases.

Council Decision 30/09/02
First call for proposals: 17.12.02-25.03.03
Indicative budget: € 109 million

LSH-2002-2.1.1-4: Rare disorders of mitochondria or of nuclear organisation with broader implications for biological processes - *IP or NoE*.

LSH-2002-2.1.1-7: Rare disorders of plasma membrane transporters for aminoacids, lipids and sugars - *STREP*.
Projects and Prospects

First call for proposals: 17.12.02-25.03.03


LSH-2002-2.1.1-12: Co-ordination of rare disease research in Europe, with various stakeholders from research, SMEs and patient organisations - SSA.
First call for proposals: 17.12.02-25.03.03

LSH-2002-2.1.3-4: Rare hereditary neurological disorders: ataxias - IP.

LSH-2002-2.1.3-7: Rare monogenic neurological disorders - STREP/CA.
Second call for proposals: 15.07.03-13.11.03
Indicative budget: € 116 million

LSH-2003-2.1.1-4: Prader-Willi Syndrome: gene expression, obesity and mental health - STREP.
LSH-2003-2.1.1-5: Rare autoimmune disorders: from genes to individualised medicine - STREP.

Rare autoimmune disorders
Pathogenesis

Genes
Polymorphisms

Genomics
Proteomics
Structural studies
Functional studies

New diagnostic, prognostic and therapeutic agents
Projects and Prospects

LSH-2003-2.1.1-7: Combating rare genetic skin disorders - CA.

- genomics
- proteomics
- translational research
- clinical investigation

accessibility to new techniques and knowledge
new diagnostic agents and treatments
Projects and Prospects


- genomics
- proteomics
- metabolomics
- glycomics
- lipidomics

Improved diagnostics
+ Innovative treatments
Projects and Prospects

LSH-2003-1.2.1-1: Medicines for children - NoE.

LSH-2003-1.2.2-3: Molecular diagnostics in mitochondrial diseases - STREP.

LSH-2003-1.2.4-2: Gene therapy of inherited diseases - IP.
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## Strengthening the Foundations of ERA

| Coordination of activities | Development of research/innovation policies |
Policy-oriented research

The activities under this heading will provide, in particular, support for: [...] Community policies, namely health [...] Providing health, security and opportunity to the people of Europe. Research in this category responds to policy requirements relating, in particular, to the implementation of the European Social Agenda including future social policy issues, public health and consumer protection and the creation of an Area of Freedom, Security and Justice. It will focus on: [...] public health issues, including epidemiology contributing to disease prevention and responses to emerging rare and communicable diseases, allergies, procedures for secure blood and organ donations, non-animal test methods. [...]
Projects and Prospects

• Overall indicative budget: € 555 million.

• First call: 17 December 2002-13 March 2003 (indicative budget € 13.3 million).

Task 5: Constitution of research capacity in the field of rare diseases: to develop a network of research capacity and scientific support in the field of rare diseases research in order to address and propose solutions to research needs that may emerge from public health activities in the area of rare diseases.
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## Strengthening the Foundations of ERA

| Coordination of activities | Development of research/innovation policies |
Support for the coordination of activities

Coordination of national activities
The objective is to encourage and support initiatives undertaken by several countries, in areas of common strategic interest, to develop synergy between their existing activities through coordination of their implementation, mutual opening and mutual access to research results, as well as to define and implement joint activities. Efforts to encourage coordination activities, using a bottom-up approach, will be carried out in the whole field of science and technology, including across domains and disciplines, in areas such as:
— health: [...] rare diseases [...]
• Overall indicative budget: € 270 million.

• Call: 17 December 2002.

• First closure date: 3 June 2003 (indicative budget € 24 million).
Next steps:

1/ negotiation of successfully evaluated proposals of 1st call

2/ preparation of evaluation of 2nd call proposals
Thank you for your attention!

Cordis website: http://fp6.cordis.lu/fp6/home.cfm

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