

# **E-Rare: Networking research programmes on rare diseases in Europe**

GIS-Institut des Maladies Rares (Paris, France)  
E-Rare Coordination Unit



Support for the coordination of activities

# What is E-Rare?

## E-Rare: ERA-Net for research programmes on rare diseases

- Project funded by the European Union through FP6 Coordination action, **4 years** (start date: June 2006)
- Objective: coordinate existing national or regional research programmes on RD to:
  - Reduce fragmentation in research
  - Launch multinational joint calls and other joint initiatives
  - Eventually develop joint programmes on RD

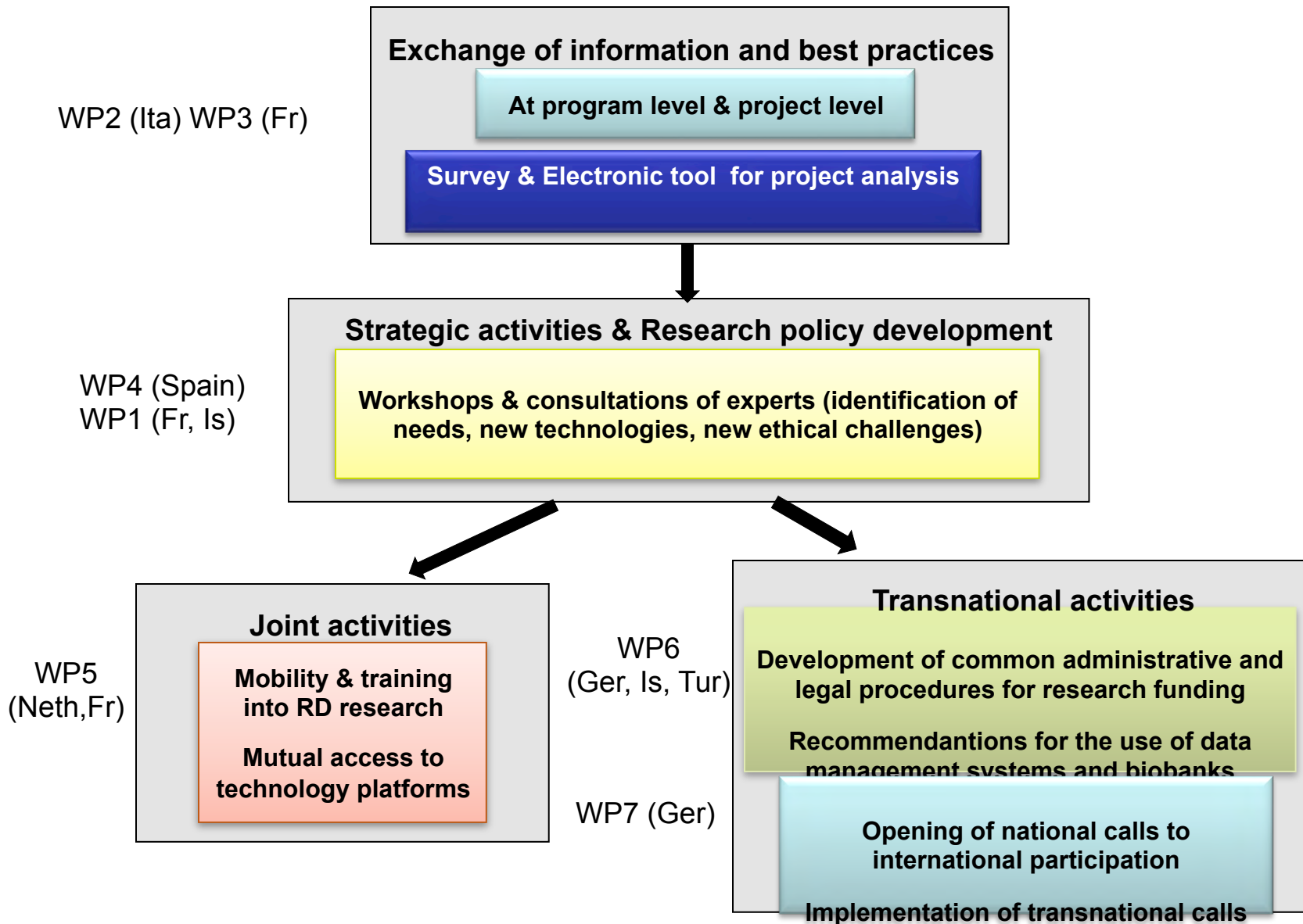


# E-Rare partners

- **France:** GIS-Institut des Maladies Rares (**coordinator**) & National Research Agency (ANR)
- **Germany:** Project Management Agency of the German Aerospace Center (PT-DLR) & Federal Ministry for Education and Research (BMBF)
- **Spain:** Institute of Health Carlos III/(FIS & IIER)
- **Belgium:** National Funds for Scientific Research (FNRS)
- **Italy:** National Institute of Health (ISS)
- **Israel:** Ministry of Health (CSO-MOH)
- **Netherlands:** Organisation for Health Research and Development (ZonMw)
- **Turkey:** Scientific and Technological Research Council (TUBITAK)

**2 affiliated partners :** Russian Ministry of Science / Region of Lombardy (Italy)

# E-Rare roadmap



# E-Rare achievements and successes (1)

## ■ **Systematic exchange of information and best practices**

- \* Expansion of mutual knowledge of partners national programmes (inventory on the state of the art on activities and national programmes)
- \* Analysis of RD projects in Orphanet's database through a web-based systemic information for project managers
- \* Establishment of a bottom-up approach in design of joint activities (e.g., JTC)/strong basis for long-term cooperation between partners

## ■ **Strategic and joint activities**

- \* WS "Database management systems and biobanks" -> information and help for researchers to scale-up quality of proposed systems
- \* WS "Opening of national programmes to more international cooperation" -> inform researchers on existing national possibilities
- \* WS "Rotational positions" -> catalogue of national possibilities/definition of unmet needs for researchers/clinicians

## **E-Rare achievements and successes (2)**

- **Launch of 2 Joint Transnational Calls (JTC) for research in RD**

- \* JTC participating countries :**

- 2007 (Fr, It, Ger, Sp, Turk, Isr) (6)
    - 2008-2009 (Fr, It, Ger, Sp, Turk, Isr, Nether, Port, Aus, Gre) (10)

- \* Launch and completion :**

- 2007 March 2007 – Nov 2007
    - 2008-2009 Dec 2008 – Oct 2009

# Scope of E-Rare Joint Transnational Calls for research in rare diseases

Epidemiology / Natural history of diseases

Registries, databases, biomarkers, diagnosis/prognosis markers

Human & Social Sciences

Genetics / Physiopathology

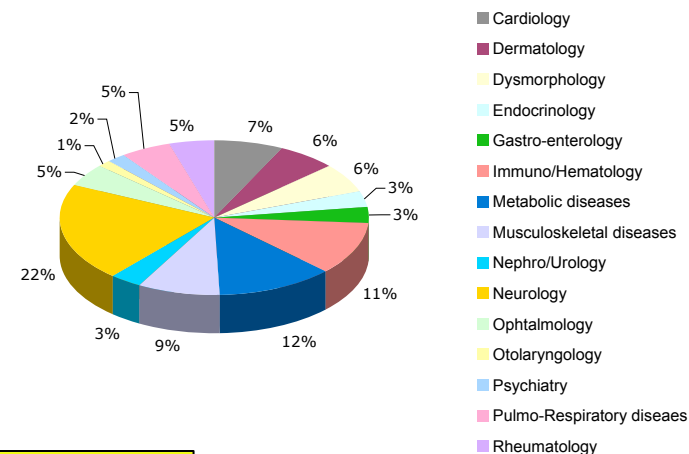
Molecular mechanisms, new animal models, omics, biomarkers, development of new therapeutic tools

Pre-/Therapeutic Research

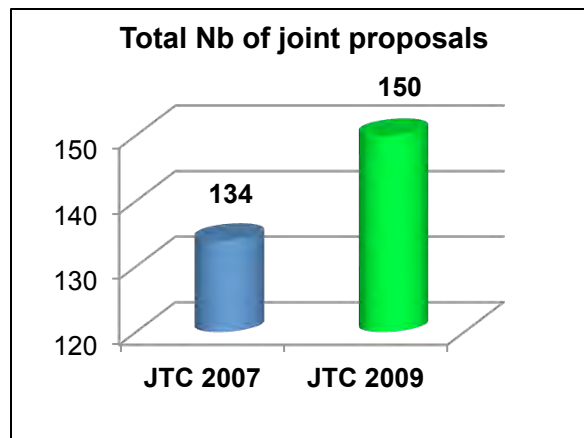
Proof-of-concept, therapeutic targets, innovative biotechn. research, drug toxico/pharmacol.

~~Cancers, rare drug effects, clinical trials~~

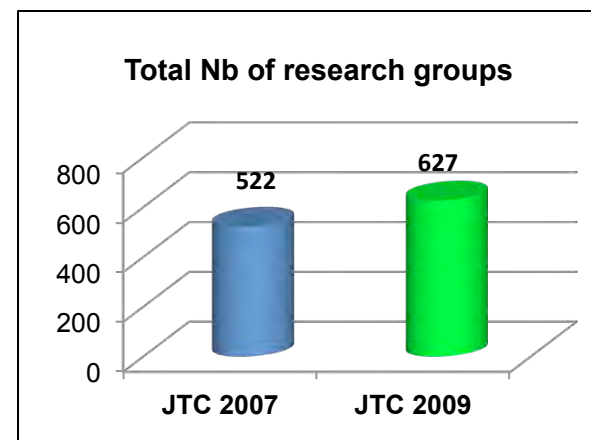
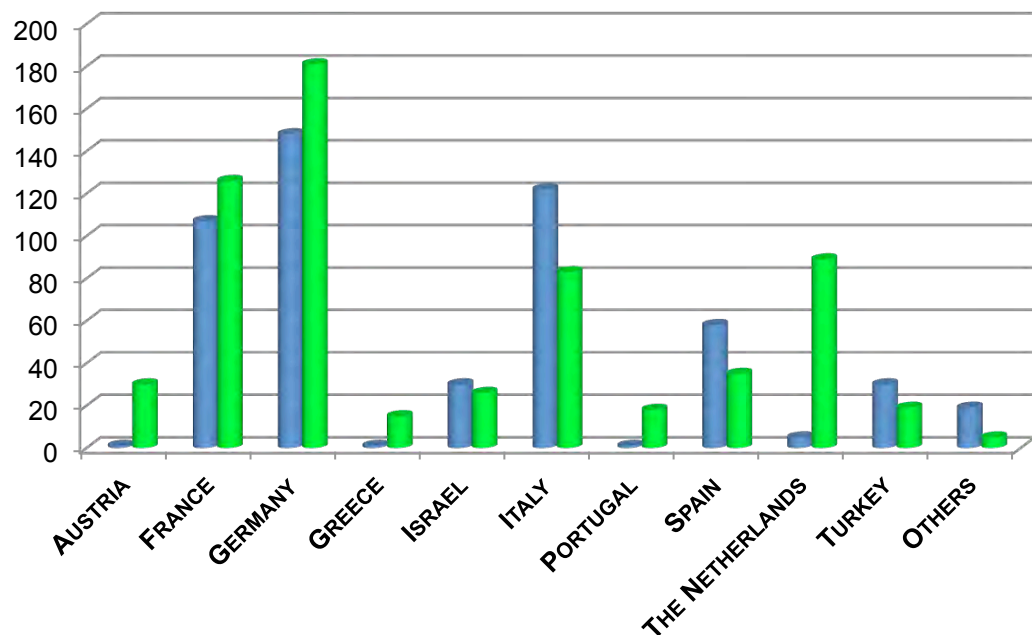
Medical domain of Submitted projects JTC 2009



# E-Rare JTCs meet the needs of the RD research community for funding joint research projects

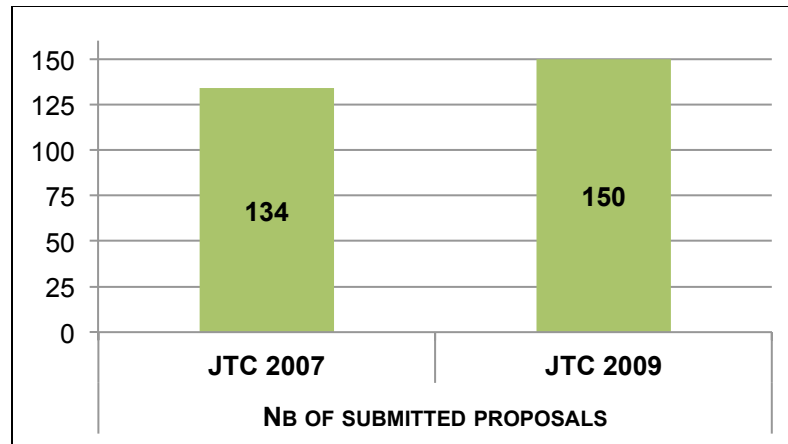


**Nb of research groups/country**

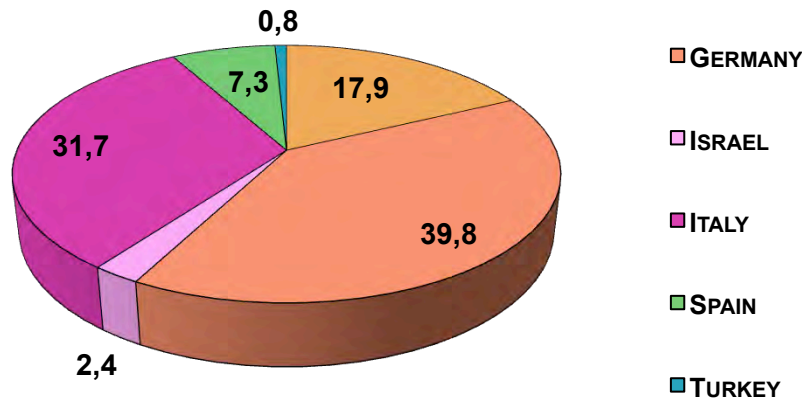




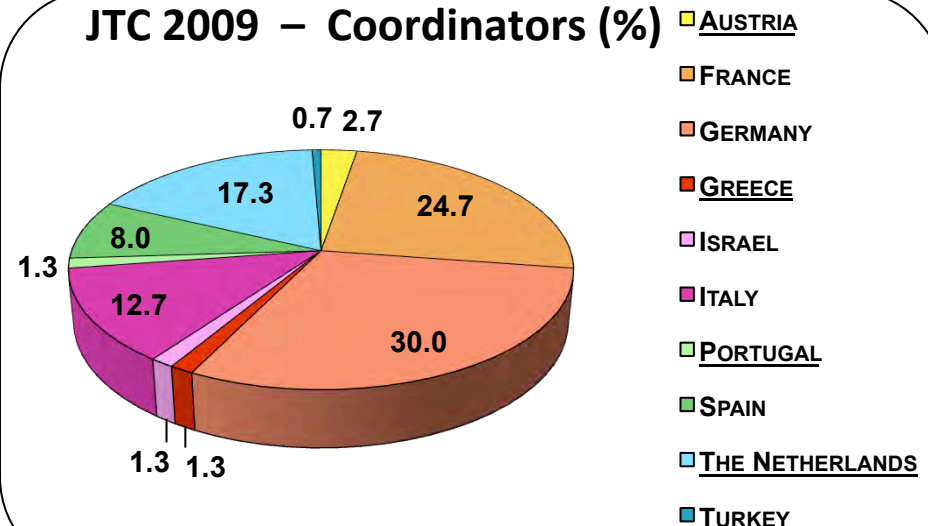
# New partner's countries take over coordination of joint transnational research proposals



JTC 2007 – Coordinators (%)



JTC 2009 – Coordinators (%)



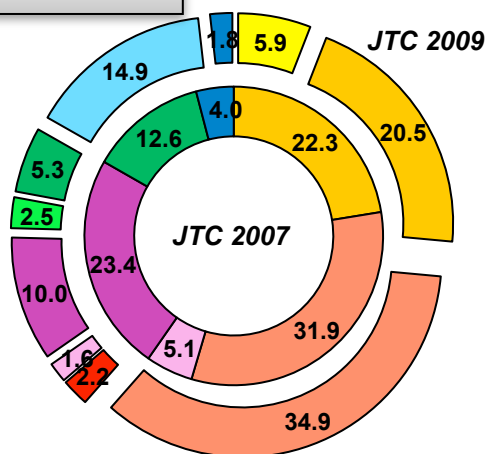
## Funds in and funding, E-Rare JTCs

- A common evaluation and ranking procedure of the proposals (International Scientific Evaluation Committee)
- A selected project is considered as a ‘whole’ and not as a summ of parties (e.g., european added-value)
- A « virtual » common pot
  - each participating country (i.e., National Funding Agencies) puts funds in the « pot »
  - each participating country finances its national research teams within selected joint projects

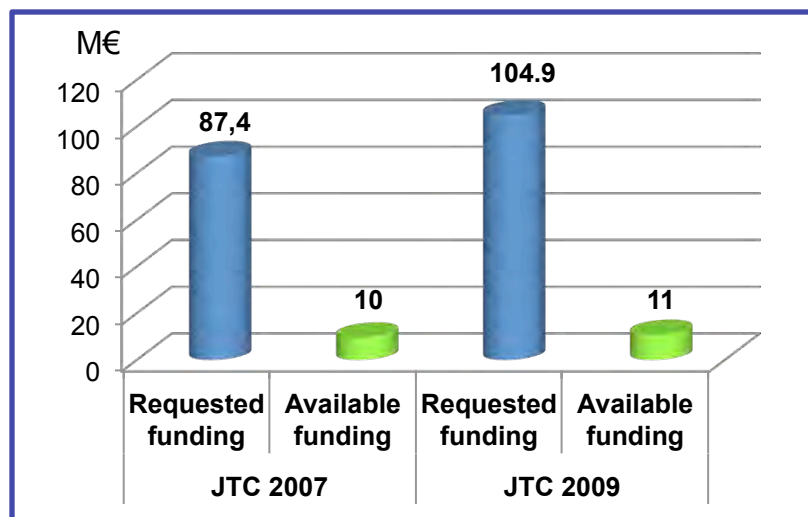
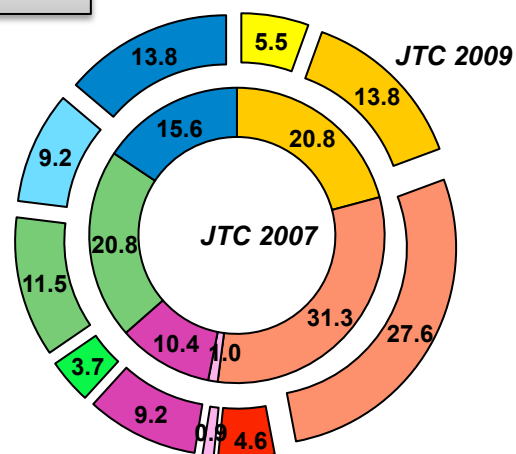
**Hence, the need to match amount of « national » funding with « national » research strength**

# E-Rare Joint Transnational Calls for research in RDs: a need for increased funding

Requested funding (%)



Available funding (%)



## **E-Rare 2006-2010 : lessons learnt**

- **Success of the E-Rare Joint Transnational Calls reflects expectations and needs of the RD research community**
- **Legal and administrative barriers among countries are still significant** (i.e., synchronisation of national calls; opening of national programmes to international collaboration...)
- **E-Rare will continue its efforts towards:**
  - Facilitating access of RD researchers to the best technology platforms regardless of their localization
  - Fostering multidisciplinary through training and exchange programmes
  - Establishing joint programmes for rare diseases research

# Continuation/Extension of E-Rare

- **Aim :**

- **develop a joint transnational RD research programme**

- **Objectives :**

- **widen European cooperation by including additional partners**
- **state of the art of research in RD across Europe**  
(identification of unmet needs, research priorities)
- **launch yearly JTC with topics more oriented on pre-therapeutical and clinical, research**
- **effective implementation of strategic joint activities** (mobility, infrastructures...)

Thank you for your attention!

[www.e-rare.eu](http://www.e-rare.eu)

A banner for ERA-NET 2006. The left side features the text 'ERA-NET' in blue and '2006' in black, with a large orange puzzle piece below. The background is a collage of colorful puzzle pieces and a close-up of a person's eye being examined with a medical instrument. The right side has a grey background with the text 'rare opportunity for co-operation' and a quote about scientific platforms and networking.

ERA-NET 2006

Coordination action

rare opportunity  
for co-operation

“ Coordination of scientific platforms and centres will open the way to networking and to the creation of a real European research programme on rare diseases. ”

## Categories des 36 RD projects funded during le FP6: Research : IIIIIII

- LSH 2002-2.1.3-4 Rare hereditary neurological disorders: ataxia<sup>1</sup> (patho, genetics, animal models..)
- LSH 2003 2.1.1-5 Rare autoimmune disorders : from genes to individualised medicine <sup>1</sup>
- LSH-2004.2.1.1-10 Pharmacogenomics related to drug targets of « designated orphan medicinal drugs » <sup>1</sup>
- LSH 2003 1.2.4-8 Use of gene transfer for curative therapy of human skin diseases <sup>1</sup>
- LSH 2002 2.1.1-7 Rare disorders of plasma membrane transporters for amino-acids, lipids and sugars <sup>1</sup>
- LSH 2002-2.1.3-7 Rare monogenic neurological disorders <sup>11</sup>
- LSH 2004-1.2.1-3 Exploring the potential of stem cells and/or primary cells, for the understanding of monogenic RD and the development of their treatment <sup>11</sup>
- LSH 2004-2.1.1-9 Rare disorders of nuclear organisation <sup>11</sup>
- LSH 2003-2.1.1-8 Combating disorders of inborn errors of metabolism <sup>1</sup>
- LSH 2003-2.1.1-7 Combating rare genetic skin disorders <sup>1</sup>

POLICIES-2.2/ Public health issues, incl.epidemiology contributing to disease prevention and responses to R and communicable diseases, allergies, procedures for secure blood and organ donation, non-animal tests methods :

LSH 2005-2.1.1-12 In vitro/animal model of rare diseases

LSH-2005-2.1.1-7: Rare inherited neuromuscular disorders: from molecular basis to cutting edge therapies - NETWORK OF EXCELLENCE

LSH-2005-2.1.1-8: Rare disorders of protein folding – STREP

LSH-2005-2.1.1-9: Rare diseases of connective tissues affecting bone and/or cartilage – STREP

### **HEALTH-2007 Natural course and history of RD 11 projects**

HEALTH-2007-1.2-6 High throughput molecular diagnostics in individual patients for genetic diseases with heterogeneous clinical presentation **2**

### **Health-2008 Rare neurological diseases**

**Pre-Clinical development of substances with a clear potential as orphan drugs & Preclinical ongoing evaluation)**

**Health-2010 Clinical development of substances with a clear potential as orphan drugs**

**+ E-Rare**



- 3rd call FP7 rare diseases **Rare neurological diseases.**  
Funding scheme: CP-FP, max. € 6m. (1 or more projects)
- **Preclinical development of substances with a clear potential as orphan drugs.**  
Funding scheme: CP-FP, max. € 3m. (1 or more projects)
- FP6 funded RD projects : 60

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