



# Rare Disease Research: Expectations and Contributions from Patients The European Workshop Outcomes

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# **Eurordis' European Workshop on « Gaining Access to Rare Disease Research Resources »**



**Institut Pasteur  
Paris  
4-5 May 2007**

Science and Society



# **Eurordis European Workshop on Gaining Access to Rare Disease Research Resources**

## **Key information:**

- 2 days: 2 plenary sessions + 2 parallel sessions**
- 270 participants**
  - 75% rare disease patient representatives,**
  - 25% researchers, healthcare professionals, industry representatives and public policy makers**
- 27 countries**

**Part of the CAPOIRA project (n°2006-044665)**

***Capacity Building Activities for Patient Organisations in Research***  
**DG-Research – Science and Society**

# **Eurordis European Workshop on Gaining Access to Rare Disease Research Resources**

## **UNDERSTANDING RARE DISEASE RESEARCH RESOURCES**

*Plenary session 4 May*

### **Rare Disease Research – A decade of progress**

**Elisabeth Tournier-Lasserre, *Director GIS- Institut des maladies rares (France)***

### **A step forward for rare disease research in Europe**

**Manuel Hallen, *Head of Unit – Medical & Public Health Research - DG Research, European Commission (EU)***

### **Capacity building for patient organisations in EU Research Activities**

**Yann Le Cam, *CEO - European Organisation for Rare Diseases, Eurordis (EU)***

### **Successful collaboration for research development between the NIH and patient organisations**

**Steve Groft, *Director, Office of Rare Diseases, National Institutes for Health (USA)***

### **Patient driven research at EU level: 3 success stories**

**Anthony Holland -“The Prader Willi European Project”**

**Alexis Arzimanoglou -“The European Network for Research on Alternating Hemiplegia”**

**Volker Straub - “The TREAT-NMD Network of Excellence”**

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**5 May**

## **How to better integrate patient organisations in research**

### **Possible modes of participation of civil society in EU research**

Viviane Willis-Mazzichi, DG Research, European Commission (EU)

### **Financing health research through the EU Framework Programmes: processes & opportunities**

Catherine Berens “Medical & Public Health Research” , DG Research,  
European Commission (EU)

**Panel debate:** Terkel Andersen, Catherine Berens, Yann le Cam,  
Christian Cottet (AFM, France), V. Willis-Mazzichi

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## **Workshop 1**

### **European tools for rare disease research**

***Thanks to European funding, several international initiatives have been set up to specifically support rare disease research.***

#### **E-Rare: recent developments in collaboration between Member States**

Pascale Borensztein, E-Rare Coordinator (FR)

#### **Building clinical research infrastructures for rare diseases**

Christian Glud, ECRIN (DK)

#### **EuroBioBank and its importance for rare disease patients**

Hanns Lochmüller, Friedrich-Baur-Institut (GE)

#### **Benefits & pitfalls of a patient database**

Jeanne-Hélène di Donato, 3 C-R Consulting

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## Workshop 2

### Getting involved in research

*The patients' perspective and experience in strategy development for research on their disease and EU research projects.*

**How can patient organisations trigger an EU funded rare disease project:**

- **The Prader Willi EU project**  
Christel Nourissier, Prader Willi (FR)
- **The European Network for Research on Alternating Hemiplegia** Tsveta Schyns  
ENRAH, (AU)
- **The TREAT-NMD Network** Peter Streng,  
Dutch Association for NMD (NL)

### **Patient Organisations as initiators of research- SMA case studies**

- Joseph Irwin, Jennifer Trust (UK)
- Monica Breitman, Fundame: Fundación española de Atrofia Muscular Espinal & Associació Catalana de Malalties Neuromuscular (SP)

**Participants were asked to address the following questions:**

- What are the basic requirements and tools that a patient organisation should have to best contribute to research on its disease?**
- What can patient representatives contribute to European research projects?**
- How and which structures and context can patient representatives be involved in research strategy?**

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## **Main recommendations:**

- **Promote national plans for rare diseases (including research policy) in Member States**
- **Raise awareness on rare disease research amongst EU decision makers**
- **Provide more opportunities for patients and EU decision makers to work together**
- **Establish EU-wide committees for rare diseases rather than national ones**
- **Keep providing information in national languages**
- **Develop new tools to facilitate debate and exchange of information and to promote joint research projects between lay patients and scientists.**

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## **Main recommendations:**

- **Disseminate information on research resources possibilities in the EU; Allow core funding for patient organisations;**
- **Create longer- term support for research projects with long term aims (e.g. projects on natural history of the disease);**
- **Develop more research projects on patient quality of life and on social impact of the disease;**
- **Simplify the procedure for getting support during the preparatory phase of research project;**
- **Implement a two-step approach: expression of interest first, and full application when pre-selected;**
- **Publish a who's who of the European Commission, with phone numbers and email addresses;**

# **Eurordis European Workshop**

## **Recommendations to increase the quality** **and the number of projects :**

- Create a permanent advisory/support desk at European level, dedicated to rare disease project development;**
- Organise training sessions on constraints of research, drug development process, clinical trials and related issues for patient representatives.**
- Calls for proposals: provide help at EU level; develop tools to help patient groups organise their own international calls for proposal**

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## **Specific Recommendations:**

**Rare disease biobanks: use EuroBioBank as a starting point for the creation of new high-quality biobanks; make researchers using biobanks understand that they need to share results with patients**

**Patient databases and registries: organise training sessions for patient representatives; write and disseminate EU guidelines on the creation of databases; develop databases linking genotypes and phenotypes that can be operated or supervised by patient groups with the support of specialists**

**Clinical trials: create a EU centralised registry of clinical trials and publish anonymised results; organise training sessions on protocols, methodology and analysis for patient representatives; ensure that results of clinical trials are fully used by the research community**

# Why should Patient Organisations be involved in Research at EU level

Rare Disease patient groups believe they have an expertise in :

- natural history of Rare Diseases
- constitution of cohorts
- biological samples donation campaigns
- reaching out to patients

They must play an active role as partners in Research Networks:

- To encourage patient-oriented research aiming at improving quality of life and life expectancy, reducing financial and social burdens
- To ensure rapid dissemination of results to patients, health professionals and scientific community.
- To make innovative research goals clear to all EU citizens

# Eurordis Position on Research Priorities for Rare Diseases

**Patients can really make the difference  
for research on rare diseases to be  
success!!**

***Thank you!***