

# Biomedical Network Research Centre on Rare Diseases

## A model of cooperative research on Rare Diseases

### *VI International Conference on Rare Diseases and Orphan Drugs* **ICORD 2010**



*March 18th – 20th, 2010*  
*Buenos Aires - Argentina*

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## What is CIBERER?

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What is CIBERER?

# Introduction



The **Biomedical Network Research Centre on Rare Diseases (CIBERER)** is one of the nine public consortiums set up at the initiative of the Carlos III Health Institute (ISCIII) to act as a reference to **coordinate and foster research** into Rare Diseases in Spain.

The centre groups clinical and basic researchers promoting **Cooperative Research**. The knowledge generated is translated from the laboratory to clinical practice, and vice versa: **Translational Research**.

CIBERER provides strategic coordination, human resources and materials as well as a collaborative environment where synergy proper to high multidisciplinary and complementary research potential can be developed.

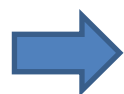


What is CIBERER?

# Objectives



- To generate knowledge,
- To translate knowledge from biomedical research into clinical practice



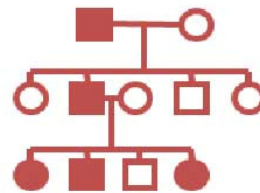
## GLOBAL CONCEPT OF THE TRANSLATIONAL RESEARCH

The scientific knowledge generated by the groups in CIBERER is intended to be applied in clinical practice in the **patients' benefit.**

Open structure to **international collaborations**, with the aim to be an international reference centre for research into the causes and mechanisms underlying rare diseases.

What is CIBERER?

# Objectives



**Benefit of Patients**



**Clinical Research**  
(Hospital)



**Translational  
Research**



**Basic Biomedical Research**  
(University, Research Centre)

**Interaction**

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# Internal structure

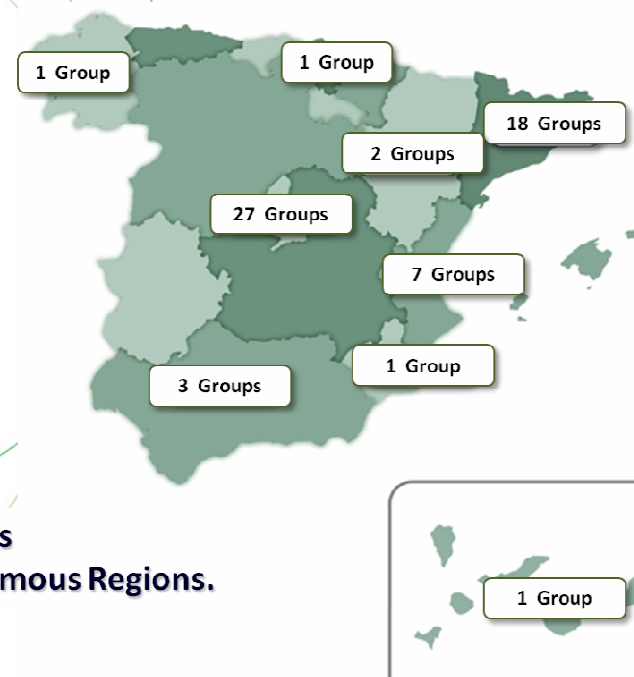


Work is made up of **61 Research Groups** below

The diagram illustrates the organizational structure of the Institute of Health Sciences (IICS). At the top, a text box states 'Work is made up of 61 Research Groups below'. Below this, a large, light blue, abstract shape represents the overall structure. This shape is composed of several overlapping, curved lines that form a complex, interconnected network. At the bottom of this network, there is a small, dark blue rectangular box containing the text '1 Group'. The IICS logo, consisting of the letters 'iics' in a stylized font with a small orange square above the 'i', is positioned in the lower right corner of the diagram. Below the logo, the text 'Instituto Argentino de Ciencias de la Salud' is written in a smaller font.



**61 Groups**  
**9 Autonomous Regions.**



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What is CIBERER?

# Internal structure



CIBERER units are organized into **five Scientific Areas**:

- **Mitochondrial Pathology** (8 research groups)
- **Inherited Metabolic and Endocrine Disorders** (20 research groups)
- **Neurogenetics** (11 research groups)
- **Clinical Genetics and Birth Defects** (15 research groups)
- **Genetic Instability and Predisposition to Cancer** (7 research groups)

The Scientific Areas are not isolated units, research lines are generally shared with other groups and collaborations are generated both within and among the different Scientific Areas.



What is CIBERER?

# Internal structure



## II. Scientific meeting for CIBERER Group leaders (Granada) 2008-04-17

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## CIBERER Programmes & Platforms

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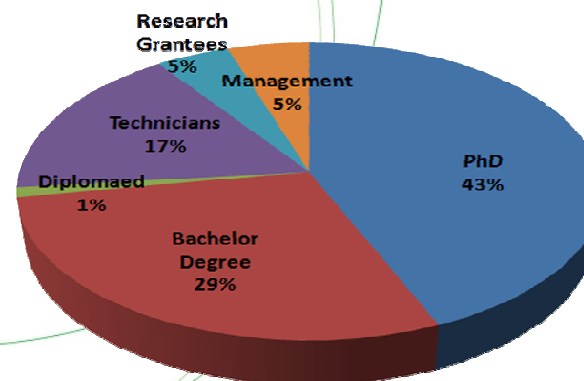
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# Human Resources Programme

To provide all the groups with a solid and reliable scientific and technical team of working professionals.

The CIBERER has large human resources - **over 700 persons**, made up of its own staff of **one hundred and ninety five CIBERER** contracted researchers, the rest of the members of the groups being personnel attached to CIBERER.

Professional category (21/12/2009)	Contracts number
PhD	85
Bachelor Degree	56
Diplomaed	2
Technicians	33
Research Grantees	9
Management	10
<b>TOTAL</b>	<b>195</b>



➡ **43% of our personnel are PhD. This reinforces and guarantees excellent research.**

# Intramural Research Projects Programme

To foster **new research lines** in the field of the knowledge concerning Rare Diseases.

Some of the Rare Diseases in study by this programme are: Friedreich's ataxia, Wolfman síndrome, Huntington's disease, Gaucher disease, Lafora disease, Fanconi's Anaemia, Congenital dyskeratosis, Hirschsprung's disease, etc.

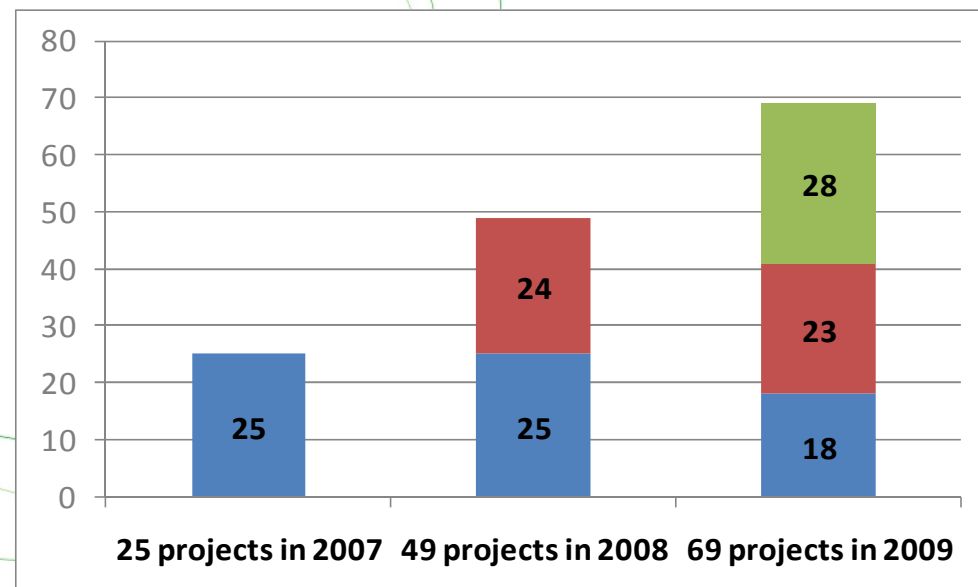
## -25 projects initiated in 2007

## -49 projects in 2008

- . Approval of 24 new projects
- . Renewal of 25 projects initiated in 2007

## -69 projects in 2009

- . Approval of 28 new projects
- . Renewal of 18 projects initiated in 2007
- . Renewal of 23 projects initiated in 2008



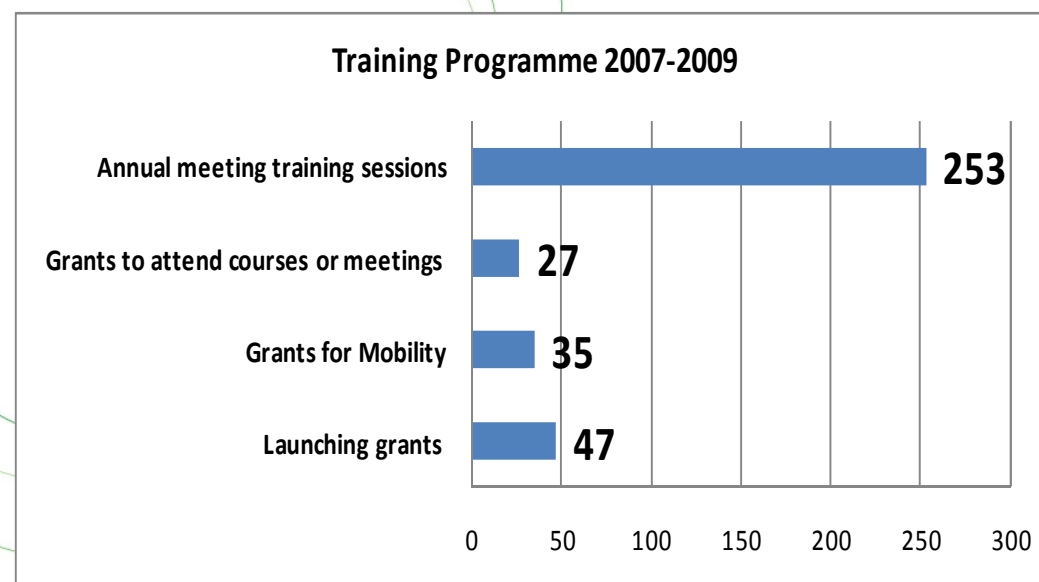


# Training Programme

To foster and supports **training activities** arranging and facilitating courses and related events.

It also encourages knowledge-sharing and cooperation between different CIBERER groups.

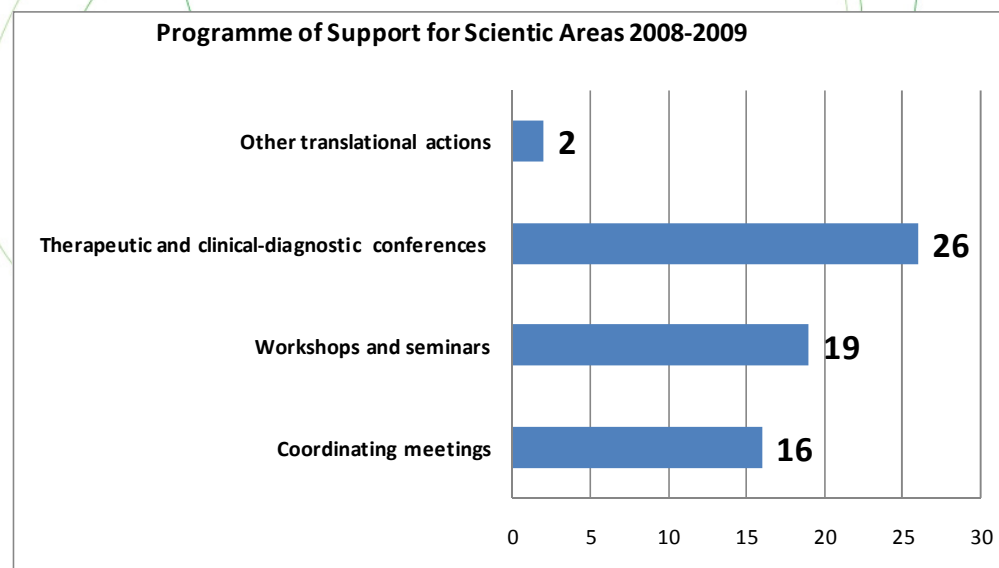
- Launching grants
- Grants for Mobility
- Grants to attend courses or meetings
- Annual meeting training sessions



# Programme of Support for Scientific Areas

To promote workshops, seminars, and above all therapeutic conferences and clinical-diagnostic conferences, covering aspects of the treatment of specific diseases at which **basic and clinical researchers**, and **patients** connected with the subject are asked to take part.

Other specific types of measures have been undertaken too, such as meetings for coordinating the areas and Internationalisation actions.



# Programme of Support for Scientific Areas

An example of therapeutic conference:

## The First National Epidermolysis Bullosa day

arranged by Dr. del Río (U714)

14th January 2009 , Hospital Niño Jesús, Madrid, Spain



For the first time in Spain, CIBERER brings together a **multidisciplinary team of experts** from around the country and **patients** affected by Epidermolysis Bullosa (DEBRA-Spain) for the treatment of this disease in the same day and at the same hospital.



Traslación



Patients



Comunidad de Madrid



## ***Día Mundial de las Enfermedades Raras*** ***II Jornada “Investigar es Avanzar”***



26th february, 2010, at the Centro Nacional de Investigaciones Cardiovasculares (CNIC), Madrid

**Objective:** To **traslate** the research work CIBERER does to society and especially to the **affected**. CIBERER involved researchers, members of patient associations and representatives of the biotechnological industry.



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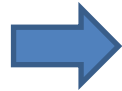


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CIBERER BIOBANK (CBK) is a public, non-profit-making biobank initiated by the Biomedical Network Research Centre for Rare Diseases (CIBERER), located in Valencia's La Fe Hospital.

With the aim of serving diagnostic and therapeutic research, by facilitating the availability of **high-quality biological material** available for research groups anywhere in the world.



**Can reduce the number of cases required to carry out research into Rare Diseases.**

[www.ciberer-biobank.es](http://www.ciberer-biobank.es)

cibererbiobank

BANCO DE MUESTRAS BIOLÓGICAS  
DE ENFERMEDADES RARAS



SEFALer is a service coordinated by CIBERER through several of its research groups unprecedented nationwide and specialize in the phenotyping of **animal models** with specific application for **Rare Diseases**.

To contribute to the study of the pathophysiology, understanding the underlying molecular mechanisms, to identify diagnostic criteria and the evaluation and refinement of new therapies.

[www.ciberer.es](http://www.ciberer.es)





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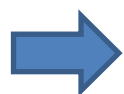
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# CIBERER Programmes

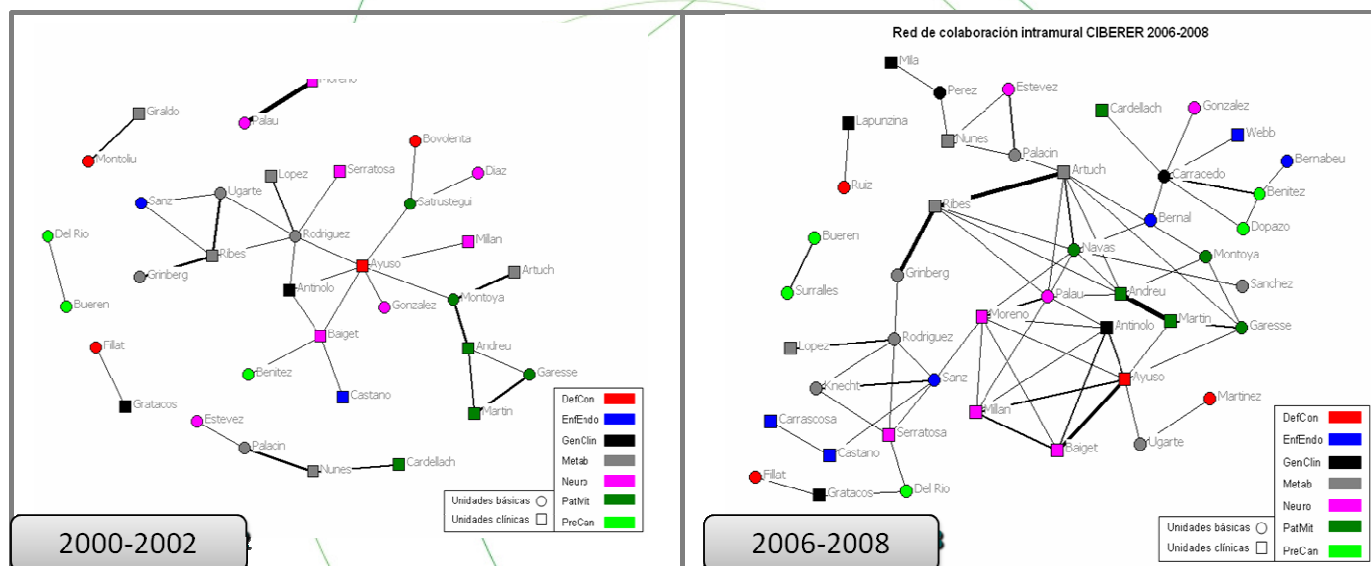
During its 3 years of existence, the CIBERER has allowed **increasing the research capacity of its scientific groups** and has increased the **knowledge about Rare Diseases**. CIBERER Programmes have:

- Joined and increased the **Cooperation** between basic and clinical units
- Increased the **Research** on Rare Diseases
- Fostered the **Participation** of groups in the CIBERER Programmes
- Established **Institutional Relations**
- **Promoted** and **Disseminated** the activities of groups



**CIBERER PROGRAMMES ARE PROMOTING TRASLATIONAL AND COOPERATIVE RESEARCH**



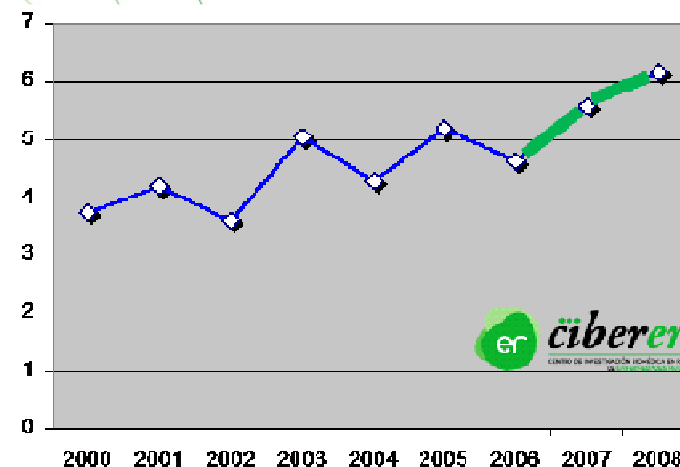


**Cluster collaboration of 61 groups, joint publications with other groups CIBERER**



NOWADAYS, THE 90% OF RARE DISEASES PUBLICATIONS IN SPAIN BELONG TO CIBERER RESEARCH GROUPS

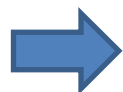
- **400** (almost 75%) of the 534 publications of the CIBERER groups in 2008 are **original works**.
- Over **37 %** of the works that were published in 2008 were in **first quartile journals**.



Increase of scientific production  
534 publications in 2008

The Orphanet logo is displayed in a blue, lowercase, sans-serif font. It is centered within a large, faint green circle that also encompasses the text below it.

The CIBERER is working closely with Orphanet, the largest database of Rare Diseases in Europe.



**Starting April 2010, CIBERER will become the Spanish partner of Orphanet.**

This collaboration intends to consolidate Orphanet as the database that unifies all the RD resources in Spain.



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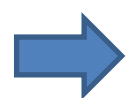
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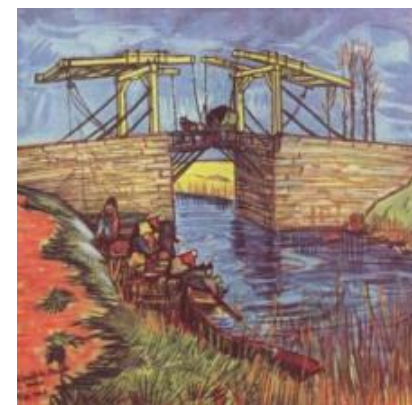


# Conclusions

CIBERER opens roads and builds bridges between researchers, doctors, institutions and patients to advance into the knowledge and treatment of Rare Diseases.



**The CIBERER is a model of cooperative research on Rare Diseases.**





**ciberer**

CENTRO DE INVESTIGACIÓN BIOMÉDICA EN RED  
DE ENFERMEDADES RARAS

## ***“A model of Cooperative Research on Rare Diseases”***

***Thank you very much for  
your attention!***

*More Information:*

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