

# Spanish Rare Diseases Registries Research Network - **SpainRDR**

<https://spainrdr.isciii.es>

Institute of Rare Diseases Research (IER)  
Instituto de Salud Carlos III

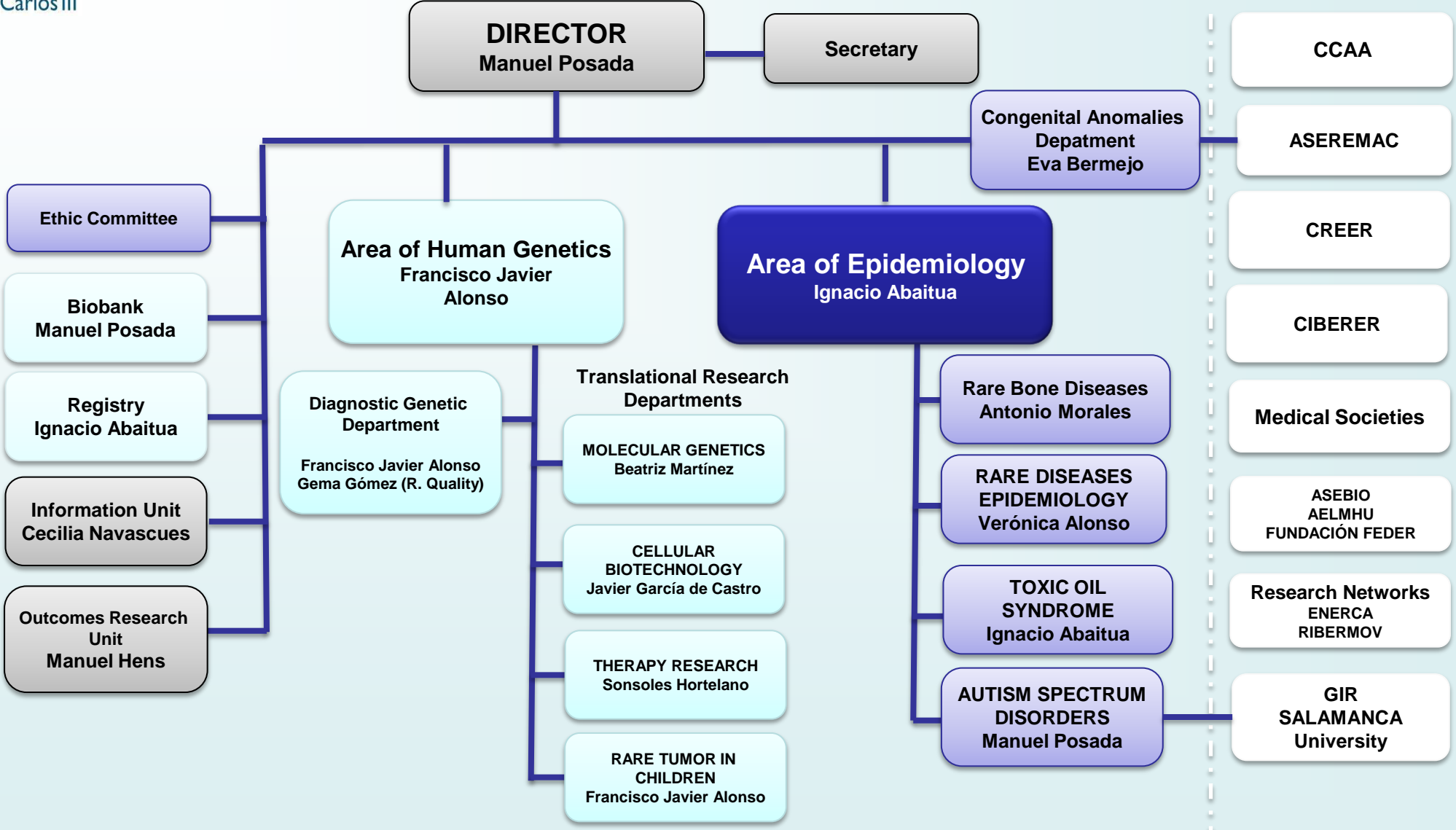
PI: Manuel Posada. Director. IIER



The Instituto de Salud Carlos III (National Institute of Health Carlos III, ISCIII) is the main Public Research Entity funding, managing and carrying out biomedical research in Spain

# Institute of Rare Diseases Research

## External Agreements



# International Rare Diseases Research Consortium (IRDiRC)



- **Worldwide action**
  - **Europe and USA initiative**
  - **Cooperation (2020)**
    - **Governance**
    - **Common data interchange**
  - **Aims**
    - **All diseases can have diagnosis**
    - **200 new RD will have therapies**
- [http://ec.europa.eu/research/health/medical-research/rare-diseases/documents\\_en.html](http://ec.europa.eu/research/health/medical-research/rare-diseases/documents_en.html)

A to Z | Sitemap | Search | About this site | Contact | Legal notice | English (en)

European Commission  
RESEARCH & INNOVATION  
Health

European Commission > Research & Innovation > Health > Medical Research > Rare Diseases

**Documents**

- Template Letter of Intent
- IRDiRC governance structure - amended version based on the outcome of the Executive Committee meeting on 25-26 January 2012
- Executive Committee meeting, 25-26 September 2012, Evry, France - Executive Summary Report
- Executive Committee meeting, 25-26 January 2012, Brussels, Belgium - Executive Summary Report
- Montreal Workshop 8-9 October 2011, documents available [here](#)
- Bethesda Workshop 5-8 April 2011 summary report (see more information [here](#))
- Reykjavik Workshop 27-28 October 2010 summary report (see more information [here](#))
- Introduction to IRDiRC

Second Interim Executive Committee meeting, 25-26 September 2012, Evry, France.

# Other Related International Actions

## RD-CONNECT

### ▶ RARE DISEASE CONNECT (RD-Connect)

RD-Connect is an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research.

[Visit RD-Connect >](#)



### ▶ EUROPEAN PLATFORM FOR RARE DISEASES REGISTRIES (EPIRARE)

The aim of EpiRare is to prepare a European platform for the registration of rare disease patients and to ensure the quality and best use of the registered data. Patients, researchers, policymakers, experts in health technology assessment and healthcare management will collaborate to build consensus and synergies to ensure success and long term sustainability of the platform.

[Visit EPIRARE >](#)

## Global Rare Disease Registry

### ▶ GLOBAL RARE DISEASE REGISTRY (GRDR)

GRDR provides a resource to the rare disease community by collecting the pan-disorder information needed to accelerate research and therapeutic advances.

[Visit GRDR >](#)

# SpainRDR project

**NATIONAL RARE DISEASES  
REGISTRY**



**HARMONIZATION**

# SpainRDR: General Aim

- **To develop a National Rare Diseases Registry based on the interaction of two different methods**
  - **Population-based registries**
  - **Patient research outcomes registries**
- **to improve prevention, diagnosis, prognosis (at different levels), treatment and quality of life for RD patients and their families using high quality information**
  - **To facilitate the implementation of RD-oriented health and social policies**
  - **To promote the translational research**

# Specific Objectives

- 1. To align actions and procedures with the international RD registry strategy to be implemented by the IRDiRC***
- 2. To develop an epidemiological rare-disease information system to support Spain's official Rare Disease Strategy and health-policy decision making***
- 3. To generate standardised criteria, including a minimum data set (MDS), common definitions of their components (common data elements-CDE), a list of standard operating procedures (SOPs) and quality assessment indicators and procedures***
- 4. To improve knowledge of RD classification and coding systems at a the Spanish national health and social services level***
- 5. To define criteria for selecting a priority RD list for promoting the inclusion of rare disease patient registries within the National RD Registry structure***



# Participants

- ❑ **Coordinator:** *Institute of Rare Diseases Research (IIER, ISCIII) Manuel Posada de la Paz*
- ❑ Spanish Ministry of Health, Centre of Reference for RD (CREER), IMSERSO
- ❑ Regional Health Departments (17 Spanish regions)
- ❑ Medical societies
- ❑ CIBERER teams
- ❑ Research Networks
- ❑ Industry
- ❑ Patient advocacy groups
- ❑ Foundations



# Organizations/Patient Registers

- ***Patient Organizations***

- FEDER.
- Fundación Teletón FEDER para la Investigación en Enfermedades Raras

- ***Industry***

- Spanish Association of Biotechnology Companies ASEBIO
- Farmaindustria
- Spanish Association of Orphan and Ultra-orphan Drugs Laboratories. AELMHU

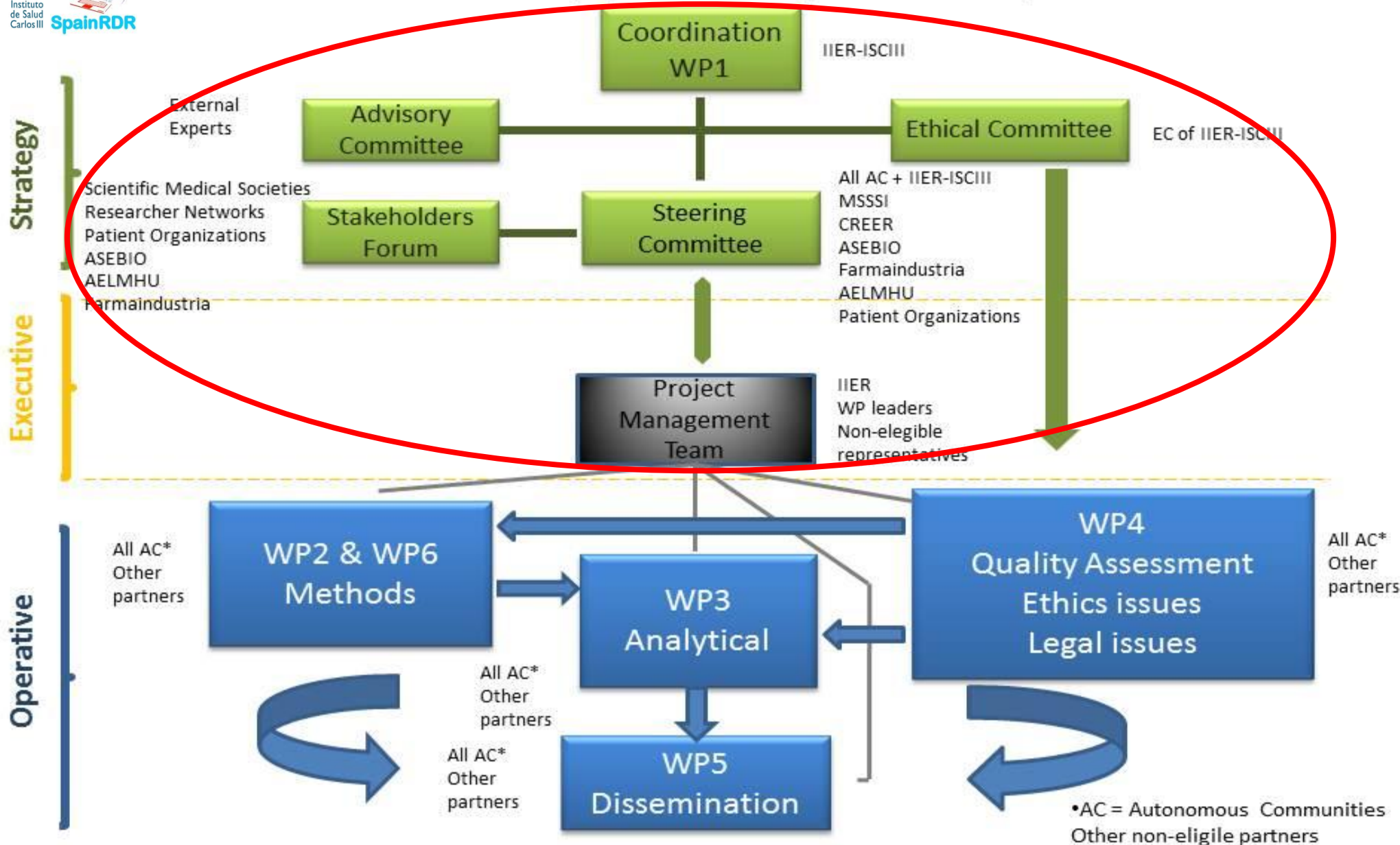
# Research Networks

- **Iberoamerican multidisciplinary network for the movement disorders study: Parkinson's Disease and Spinocerebellar Ataxias (RIBERMOV).**
- **European Reference Network for Rare and Congenital Anemias (ENERCA)**
- **Head of Regenerative Medicine Unit, CIEMAT**
- **Estudio Colaborativo Español de Malformaciones Congénitas (ECEMC) *Spanish Collaborative Study of Congenital Malformations***

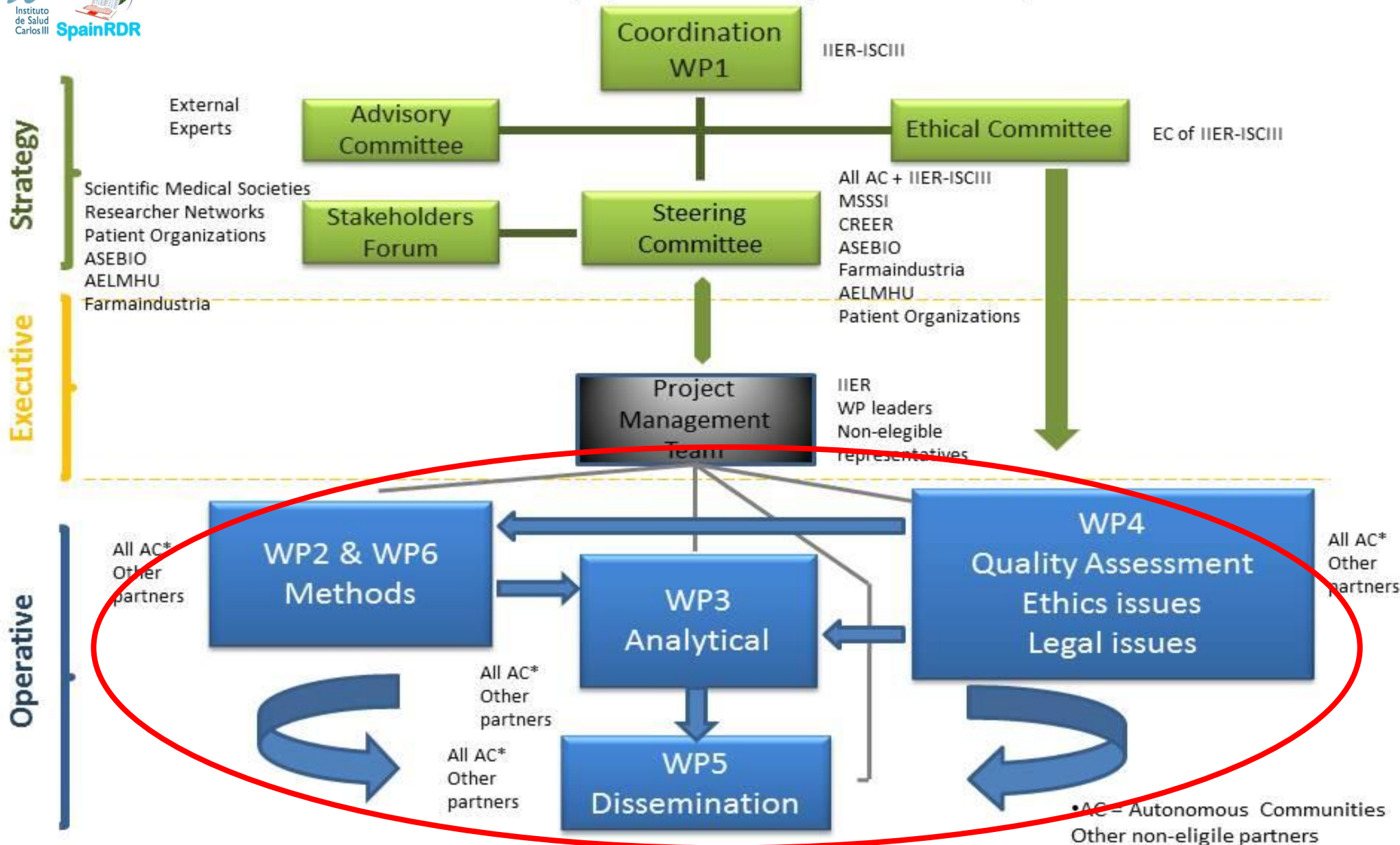
## **Agreement not signed yet**

- **The Spanish Research Group of Genetic Mental Retardation (GIRMOGEN)**
- **CIBERNED, Neuromuscular diseases**
- **European Project about McArdle's disease**

# Figure 3. Spanish Rare Diseases Registries Research Network (SpainRDR) Roadmap

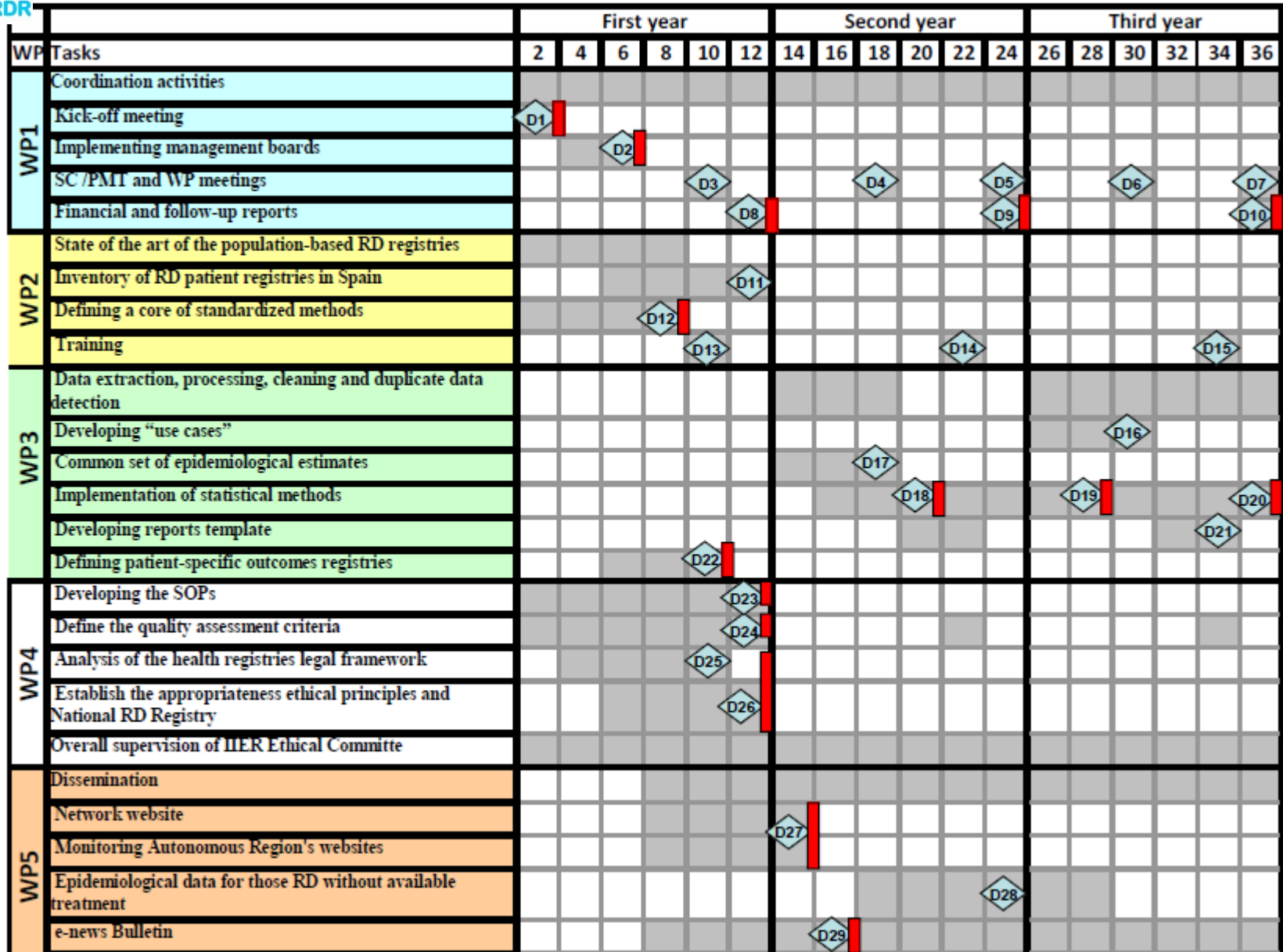


# Figure 3. Spanish Rare Diseases Registries Research Network (SpainRDR) Roadmap



**Milestones**

**D# Deliverables**

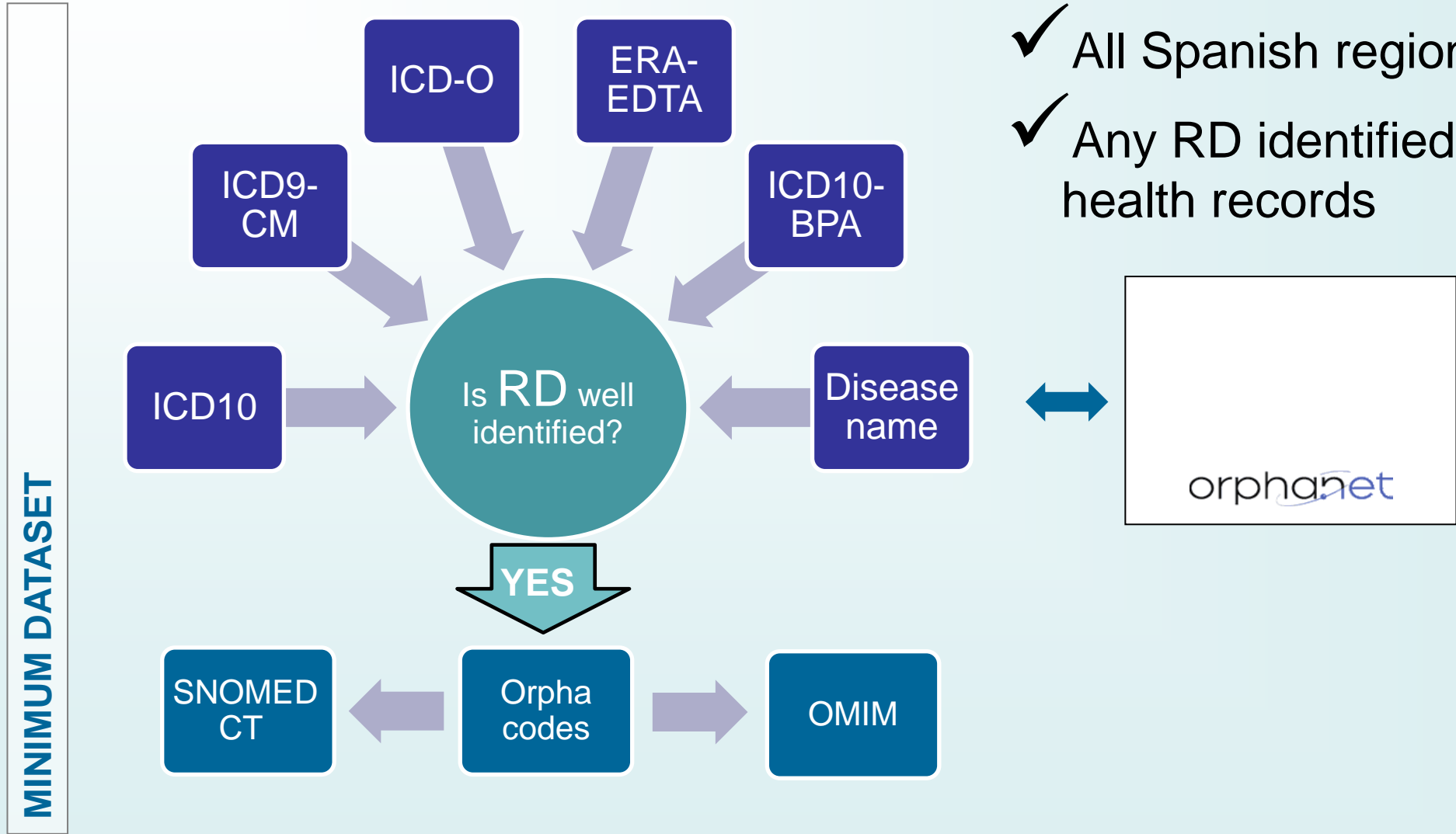


# WP 1: Advisory Board

- **Stephen Groft**, Director ORDR, NIH, USA
- **Hanns Lockmuller**, Chair of Experimental Myology, IGM, NWU- IP RD-CONNECT; Scientific Coordinator IRDiRC
- **Domenica Tarsucio**, Director CNMR, ISS; EPIRARE
- **Rumen Stefanov**, Dean, Faculty of Public Health. Medical University of Plovdiv, Bulgaria; Director, Information Centre for Rare Diseases and Orphan Drugs
- **Antoni Montserrat**, Policy Officer for Cancer, Rare Diseases and Neurodevelopmental disorders Directorate of Public Health (SANCO C-1). European Commission
- **Xavier Badia**, Senior Principal HEOR, IMS Health

# Population-based registry

- ✓ All Spanish regions
- ✓ Any RD identified in health records





**Welcome to SpainRDR!**

Welcome to the SpainRDR website. The Spanish Rare Diseases Registries Research Network-SpainRDR is a project financed by the Institute of Health Carlos III (ISCIII) within the scope of the IRDiRC for the years 2012 to 2014 with 2.4 M €.

This project involves all Health Departments of the Autonomous Communities (regions) of Spain, the Spanish Ministry of Health, the Spanish Centre of Reference of People and Families affected by RD (CREER), six Spanish Medical Societies, four research networks, pharmaceutical and biotechnological organizations (ASEBIO, AELMHU and FARMADINDUSTRIA), the Spanish Federation of RD (FEDER) and its foundation (FEDER TELETHON FOUNDATION), and the Institute of Rare Diseases Research (IER) which acts as a coordinator and leader of the network.

SpainRDR aims to build the National Rare Diseases Registry in Spain based on the input of two different strategies: patient registries addressed to patient outcome research and population-based registries addressed to epidemiologic research and social and health systems planning.

**IRDiRC**  
The International Rare Diseases Research Consortium (IRDiRC) was launched in April 2011 to foster international collaboration in rare diseases research. IRDiRC will team up researchers and organisations investing in rare diseases research in order to achieve two main objectives, namely to deliver 200 new therapies for rare diseases and means to diagnose most rare diseases by the year 2020.

**RD-Connect**  
RD-Connect is a unique global infrastructure project that links up databases, registries, biobanks and clinical bioinformatics data used in rare disease research into a central resource for researchers worldwide.

**EpiRare**  
The aim of EpiRare is to prepare a European platform for the registration of rare disease patients and to ensure the quality and best use of the registered data.

**NIH** National Center for Advancing Translational Sciences  
GLOBAL RARE DISEASES PATIENT REGISTRY DATA REPOSITORY (GRDR)  
GRDR provides a resource to the Rare Diseases community by collecting the pan-disorder information needed to accelerate research and therapeutic advances.

**News**  
News alert from GRDR pilot project  
Stephen Groft, paladín de la investigación de enfermedades raras, se jubila  
Stephen Groft, Champion of Rare Diseases Research, Retires

**Other RD Registries Actions**  
European Network for Rare and Congenital Anaemias  
The Alpha One International Registry  
Patient Registry Item Specifications and Metadata ...  
GRDR Global Rare Diseases Patient Registry and Dat...

**Events**  
10/03 2014 Workshop on orphan product EMA, London, UK  
08-10 05/2014 ECRD 2014 Berlin Berlin, Germany

**Links**  
DIG SANCO Rare Diseases  
EUCERD  
Orphanet  
Eurodis

<https://spainrdr.isciii.es>

**ONGOING**

Validation of 1,046,394 RD cases identified in health records (population-based registry)

# Bienvenido al portal de registro de enfermedades raras

Bienvenidos al portal del Registro de Enfermedades Raras del Instituto de Salud Carlos III (ISCIII), desarrollado desde el seno del Instituto de Investigación en Enfermedades Raras (IIER), centro perteneciente al ISCIII y que también forma parte del CIBERER (Consortio de Investigación Biomédica en Red de Enfermedades Raras).

Registro >>

Consultar Lista de  
Enfermedades Raras

Manual de Usuario

Preguntas Frecuentes

USUARIOS ONLINE: 26  
NÚMERO DE VISITAS: 12831

Global Repository  
<https://registroraras.isciii.es>

## Login

Usuario:   
Contraseña:

## Últimas Noticias

Enlace Externo

31/01/2013-01/03/2013

Mejorar el acceso a los medicamentos huérfanos-Revisión de la Directiva europea sobre la Transparencia

Enlace Externo

06/02/2013-06/03/2013

EURORDIS eNews 6 febrero 2013

Enlace Externo

>>ver más noticias

## Enlaces

sobre Enferdades Raras  
?

Servicio de Información y  
Orientación en ER

si@isciii.es

Tel: 902 16 17 25

>>ver más enlaces

# Data entry



Patients

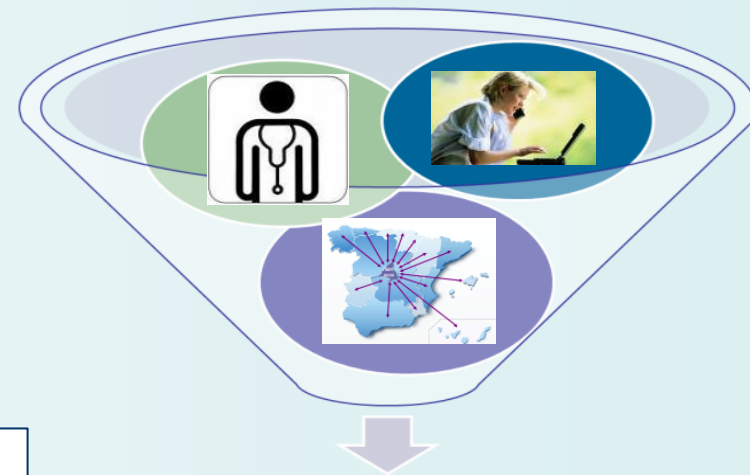


Researchers and  
Clinicians

**Patient registry**  
RESEARCH



**Population-based registry**  
HEALTH PLANNING AND POLICIES



RARE DISEASES REGISTRY



Public Health  
Departments

# General scheme for patient registered by themselves

## INSTITUTO DE SALUD CARLOS III



# National Rare Diseases Biobank and Registry

**NATIONAL RARE DISEASES REGISTRY (SpainRDR)**

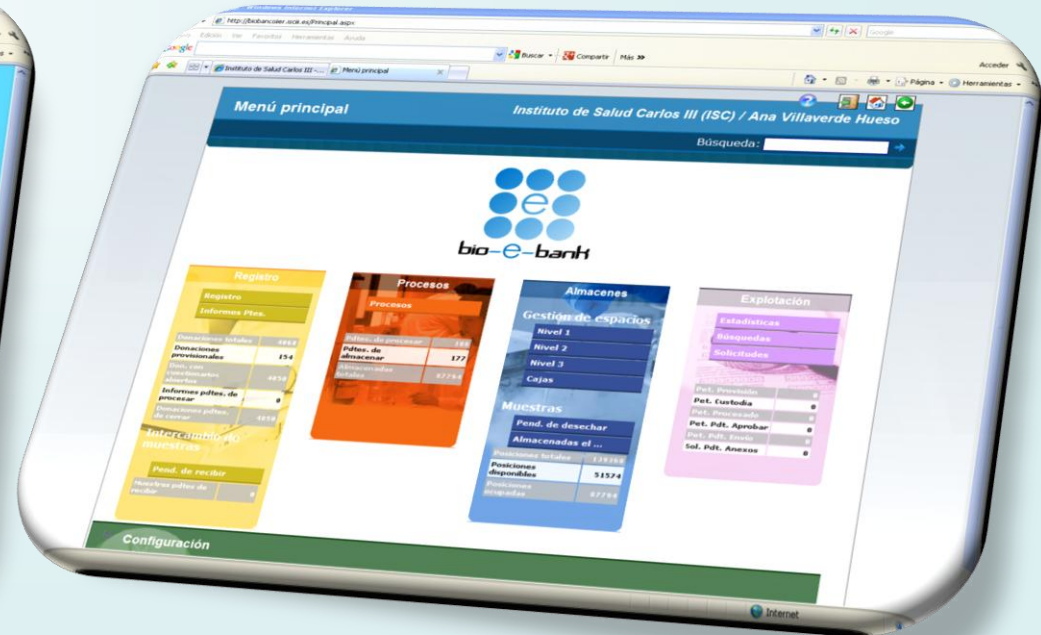
INTEROPERABLES



**RARE DISEASES NATIONAL BIOBANK (BioNER)**



<https://registroraras.isciii.es/>

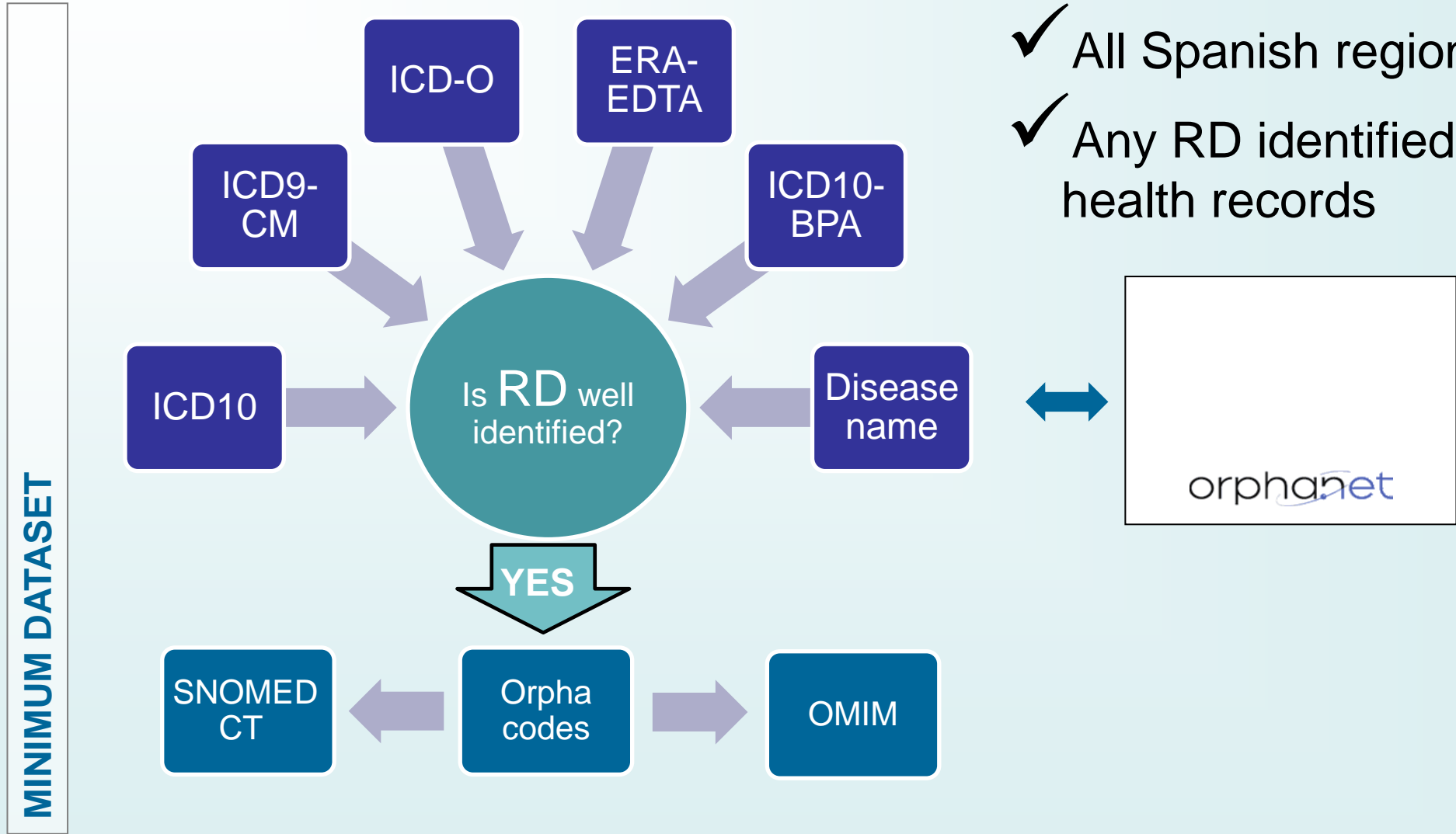


[biobanco\\_iier@isciii.es](mailto:biobanco_iier@isciii.es)

EUROBIOBANK  
 Biobank Platform, ISCIII (RetBioH)

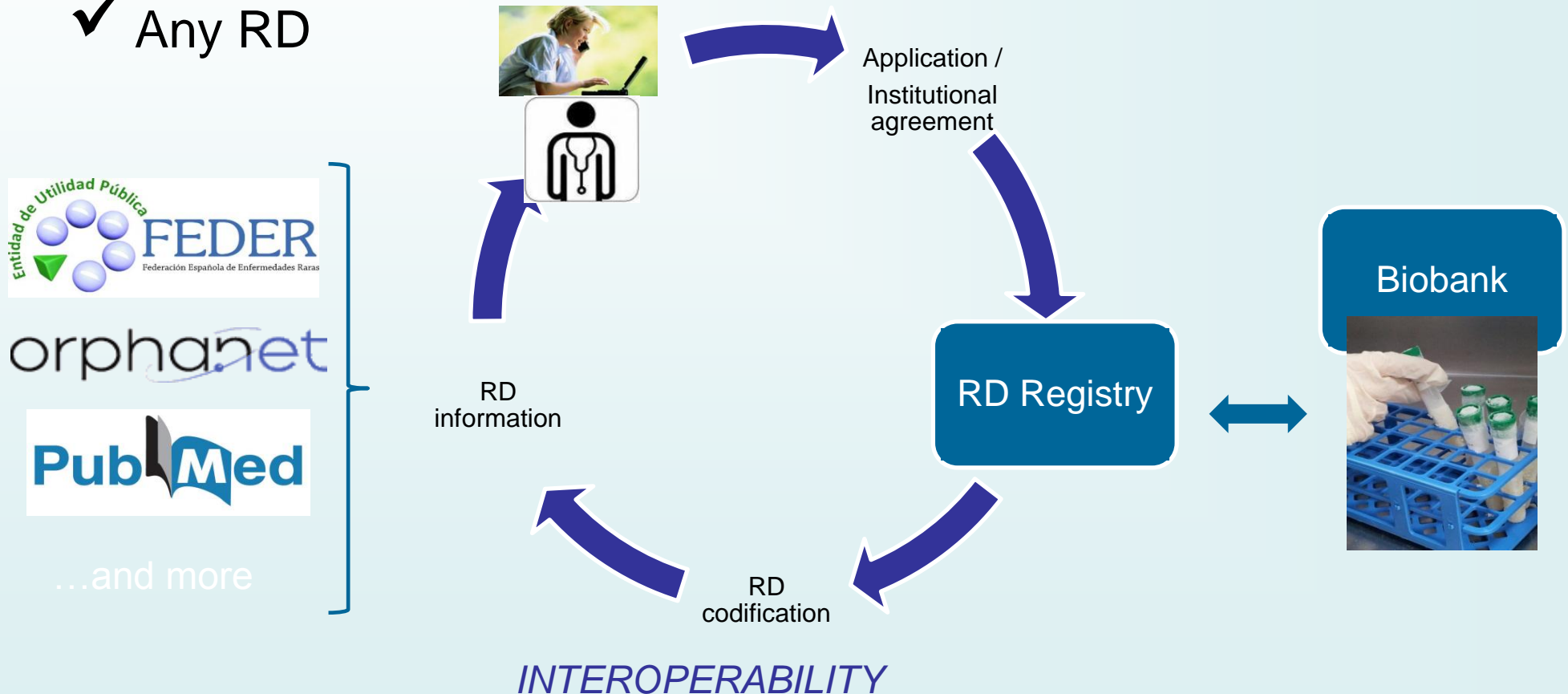
# Population-based registry

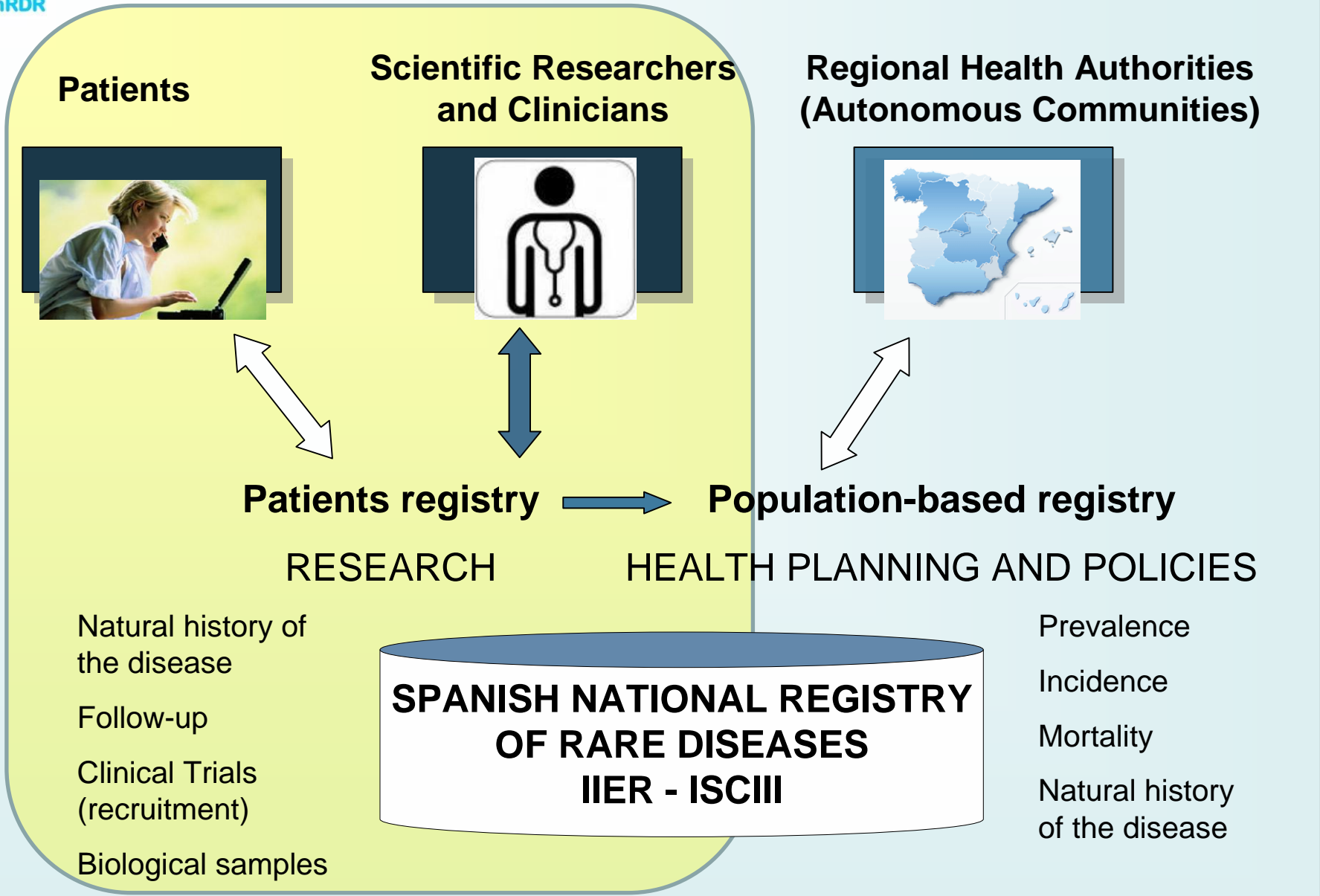
- ✓ All Spanish regions
- ✓ Any RD identified in health records



# Patient registry

- ✓ Patients, families, research networks, medical societies
- ✓ Any RD







# Medical Societies

**Spanish Society of Allergy and Clinical Immunology (SEAIC)**

**Spanish Society of Pneumology and Thoracic Surgery (SEPAR)**

**Spanish Society of Family and Community Medicine (SEMFYC)**

**Spanish Society of Pediatric Pneumology (SEPN)**

**Spanish Society of Neurology (SEN)**

**Spanish Society of Pediatric Endocrinology (SEEP)**

**Spanish Association of ASD Professionals (AETAPI)**

**Spanish Confederation of Intellectual Disability People Organizations  
(FEAPS)**

**Spanish Association of Human Genetic (AEGH)**

**Under negotiation**

Spanish Society of Pediatric Neurology (SENEP)

Developmental Neurosciences Association of Andalusia (ANDA)

Spanish Association of Neonatal Screening (AECNE)

# Patients Registries

## **SEEP**

- **Differentiation Sexual Disorders (DSD)**

## **SEAIC**

- **Bradikinin mediated angoiaedema**

## **SEPAR**

- **Alveolar proteinosis; Alpha-1 antitrypsin deficiency; Histiocytosis X; Lymphangiomatosis; Sarcoidosis; Tracheal stenosis**

## **SENP**

- **Pulmonar Interstitial Fibrosis in children**

## **SEN**

- **Spinocerebellar ataxias and Paraparesia Spastic Familiar**

## **CIEMAT**

- **Epidermolysis Bullosa**

## • **SEEP**

- **Congenital Suprarenal Hyperplasia**

## • **SEOHP+ENERCA+SEOH**

- **Congenital and rare aenemias**

## • **European Network**

- **Spanish registry of patients with McArdle disease**

## • **SEOM**

- **Rare cancers: Retinoblastoma; Ewing tumor; Suprarenal Cancer; Familiar Pancreas Cancer**

## • **FUNDAME**

- **AME**

- **All rare diseases included in the neonatal screening national program**

- **All diseases with OD marketed in Spain**

# Translational Research

- **National RD Strategy**
- **Projects**
  - **TREAT-NMD – Clinical Trial**
  - **School and RD children (bullying)**
  - **Drugs repurposing**
  - **Ictiosis**
  - **mRNA and autoimmune diseases**

# Next Steps

- **To consolidate the Population-based registry - Regions**
  - Legal Framework
  - First results
- **Going-on with the promotion of patient registries**
  - Validity
  - Audit
  - Quality Assurance Plan
- **Dissemination**
  - Websites
  - Papers
- **Alignment with other related projects**
  - GRDR and RD-CONNECT
  - European RD Platform (Ispra, Italia)
- **Ontologies and GUID**

# Summary

- **SpainRDR is covering an important role in the RD field in Spain**
  - **Full Cooperation (NHS & SS; Researchers; Industry and Patient Organizations - FEDER)**
- **Challenging topics**
  - **Case ascertainment**
  - **Validity and reliability data**
  - **Sostenibility**

**A national experiency useful for some others**

# Acknowledgements

Thank you very much  
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