Spanish Rare Diseases Registries Research Network - SpainRDR

https://spainrdr.isciii.es

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

Exp: IR11/RDR-01
Years: Dec, 2011-Dec, 2013 (2012-2014)

PI: Manuel Posada. Director. IIER

An initiative of the International Rare Diseases Research Consortium-IRDiRC
Rare Diseases National Strategy

• Development of the Strategy Lines
  – Information on Rare Diseases
  – Prevention and Early Detection
  – Healthcare
  – Therapies
  – Integrated Health and Social Care
  – Research
  – Training

SpainRDR: Objetivo General

• To set up a National Rare Disease Registry using the interaction between two different methodological approaches
  • Population-based registries
  • Patient registries outcomes research
• The overall aim is to improve prevention, diagnosis, prognosis (at different levels), treatment and quality of life for RD patients and their families using high quality information provided by the RD registry
  • To provide the necessary information to the NHS
  • To facilitate the implementation of RD-oriented health and social policies
  • To promote the translational research
SpainRDR: General Objective

• To set up a National Rare Disease Registry based on patient- and population-based registries strategies
  • IIER, ISCIII; MSSSI & CREER;
  • Regional Health Authorities (Autonomous Communities)
• Several Stakeholders
  • Medical societies
  • Research Networks
  • Industry
  • Patient organizations
  • Foundations

SpainRDR: General Objective (cont)

• To share common data
• To provide the necessary information to the NHS
• 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

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MSSSI & Autonomous Communities

- Comunidad Autónoma de Galicia
- Gobierno del Principado de Asturias
- Comunidad Autónoma de Cantabria
- Comunidad Autónoma del País Vasco
- Comunidad Foral de Navarra
- Comunidad Autónoma de Cataluña
- Comunidad de Aragón
- Comunidad Autónoma de La Rioja
- Comunidad Autónoma de Castilla-León
- Comunidad de Madrid
- Comunitat Valenciana
- Región de Murcia
- Junta de Comunidades de Castilla-La Mancha
- Comunidad Autónoma de Extremadura
- Comunidad Autónoma de Andalucía
- Illes Balears
- Comunidad Autónoma de Canarias

MSSSI (Some units)
INGESA- MSSSI
CREER-IMSERSO-MSSSI

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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

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)

*Under negotiation*
Spanish Society of Pediatric Neurology
Spanish Society of Ophthalmology
Pediatric Spanish Association or their associations
Spanish Association of Neonatal Screening (AECNE).
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 (GIRMGEN)
- CIBERNED, Neuromuscular diseases
- European Project about McArdle’s disease

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<table>
<thead>
<tr>
<th>WPs</th>
<th>Deliverables</th>
<th>Partner Leaders</th>
<th>End Month(s) Milestones</th>
<th>Month</th>
</tr>
</thead>
<tbody>
<tr>
<td>WP1</td>
<td>Co-ordination and management</td>
<td>Asturias</td>
<td>2</td>
<td>M1 2</td>
</tr>
<tr>
<td>WP2</td>
<td>Implementing management boards</td>
<td>Asturias</td>
<td>6</td>
<td>M2 6</td>
</tr>
<tr>
<td>WP3</td>
<td>Design and follow-up reports</td>
<td>Asturias</td>
<td>12</td>
<td>M6 12</td>
</tr>
<tr>
<td>WP4</td>
<td>Defining a core of standardized methods</td>
<td>Asturias</td>
<td>9</td>
<td>M3 9</td>
</tr>
<tr>
<td>WP5</td>
<td>Training</td>
<td>All Autonomous Communities</td>
<td>30</td>
<td>M5 30</td>
</tr>
<tr>
<td>WP6</td>
<td>Developing “use cases”</td>
<td>C. Valenciana</td>
<td>34</td>
<td>M4 34</td>
</tr>
<tr>
<td>WP7</td>
<td>Defining patient-specific outcomes registries</td>
<td>Asturias</td>
<td>12</td>
<td>M3 12</td>
</tr>
<tr>
<td>WP8</td>
<td>Developing the SOPs</td>
<td>Catalunya</td>
<td>12</td>
<td>M6 12</td>
</tr>
<tr>
<td>WP9</td>
<td>Analysis of the health registries legal framework</td>
<td>IIER</td>
<td>10</td>
<td>M5 10</td>
</tr>
<tr>
<td>WP10</td>
<td>Establishing the appropriateness principles and National RD Registry</td>
<td>IIER</td>
<td>12</td>
<td>M7 12</td>
</tr>
<tr>
<td>WP11</td>
<td>Dissemination and impact</td>
<td>IIER</td>
<td>16</td>
<td>M9 16</td>
</tr>
</tbody>
</table>
Leaders of WPs Teams

- WP1. Co-ordination and management – Manuel Posada
- WP2. Registering activity-related methods – Mario Margolles (Asturias)
- WP3. Data-analysis and outcomes research – Óscar Zurriaga
- WP4. Quality assessment and ethical and legal issues – Josep Jiménez
- WP5. Dissemination and impact – Manuel Posada
- WP6. Patient Registries – Manuel Posada

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
WP 1: Ethic Committee

- Javier Júdez Gutiérrez (Chair)
- Lydia Feito Grande
- Antonio Morales Piga (Secretary)
- Mª Concepción Martín Arribas
- Teresa Pámpols Ros
- Jaime Campos Castell
- Pilar Nicolás Jiménez
- Juan Fuertes Guillén (FEDER)
- Mauro Rosati (FEDER)

Patients Registries

- SEEP
  - Differentiation Sexual Disorders (DSD)
- SEAIC
  - Bradykinin mediated angioedema
- SEPAN
  - Alveolar proteinosis
  - Alpha-1 antitrypsin deficiency
  - Histiocytosis X
  - Lymphangiomatosis
  - Sarcoidosis
  - Tracheal stenosis
- SENP
  - Pulmonary Interstitial Fibrosis in children
- SEN
  - Spino-cerebellar ataxias and Paraparesia
  - Spastic Familar
- CIEMAT
  - Epidermolysis Bullosa

SEEP
- Congenital Suprarenal Hyperplasia
SEOH+ENERCA+SEOH
- Congenital and rare anemias
European Network
- Spanish registry of patients with McArdle disease
SEOM
- Rare cancers: Retinoblastoma; Ewing tumor; Suprarenal Cancer; Familiar Pancreas Cancer
- All rare diseases included in the neonatal screening national program
- All diseases with OD marketed in Spain

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Spanish National RD Registry

INSTITUTO DE SALUD CARLOS III

REGISTRO DE PACIENTES CON ENFERMEDADES Raras

SOLICITUD

ACEPTADA

PROFESIONAL

ADMINISTRACIONES

CIUDADANO

ADMINISTRADOR ENFERMEDAD RARA

Consentimiento informado
Informe diagnóstico

REGISTRO DE ENFERMEDADES Raras

CUESTIONARIOS

BIOBANCO NACIONAL DE ER

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Rare Diseases National Biobank
Europea Network of Rare Diseases Biobanks – EUROBIOBANK
National Netowork of BiobanksISCIII (RetBioH)
Training on-line

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https://spainrdr.isciii.es

https://registroraras.isciii.es
Other Related International Actions

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Thank you

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Instituto de Salud Carlos III

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