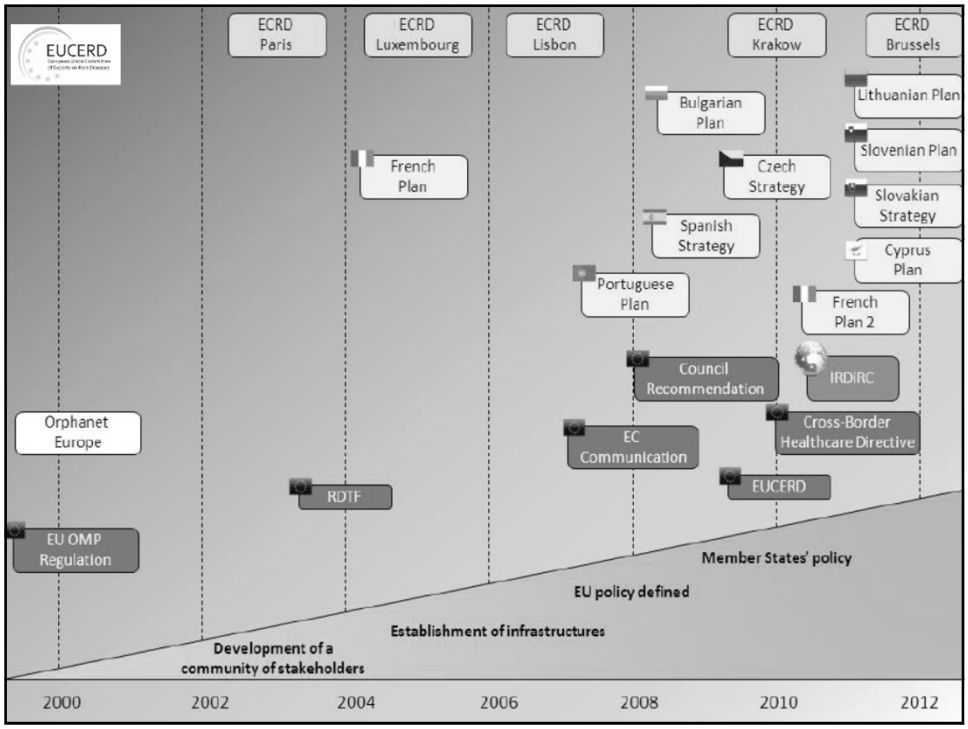
Investigación en Enfermedades Raras

JORNADA
Actualización de
Recursos de Atención
en Enfermedades Raras
Burgos 24 de junio de 2014

Manuel Posada
Director
Instituto de Investigación en Enfermedades Raras



European Strategic Framework

- ***Communication from the Commission***

COMMISSION OF THE EUROPEAN COMMUNITIES

Brussels, 11.11.2008
COM(2008) 679 final

COMMUNICATION FROM THE COMMISSION TO THE EUROPEAN PARLIAMENT, THE COUNCIL, THE EUROPEAN ECONOMIC AND SOCIAL COMMITTEE AND THE COMMITTEE OF THE REGIONS

on Rare Diseases: Europe's challenges

[SEC(2008)2713]
[SEC(2008)2712]

- ***Council recommendations***

COUNCIL OF THE EUROPEAN UNION

Brussels, 5 June 2009
(OR. en)

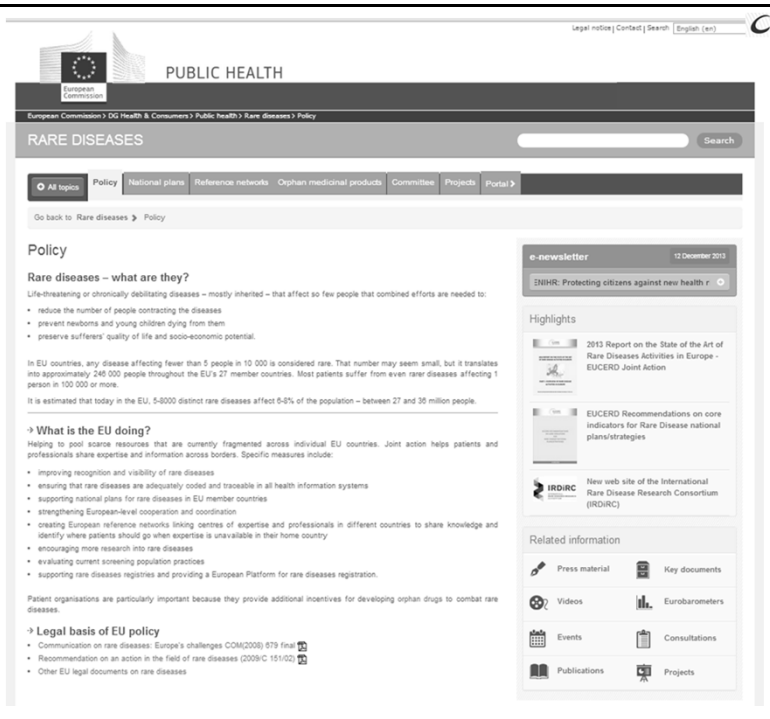
10122/09

Interinstitutional File:
2008/0218 (CNS)

LIMITE

SAN 142
RECH 167
MI 219

LEGISLATIVE ACTS AND OTHER INSTRUMENTS
Subject: COUNCIL RECOMMENDATION on an action in the field of rare diseases



Legal notice | Contact | Search English (en)

PUBLIC HEALTH

European Commission > DG Health & Consumers > Public health > Rare diseases > Policy

RARE DISEASES

Search

All topics | Policy | National plans | Reference networks | Orphan medicinal products | Committee | Projects | Portal

Go back to Rare diseases > Policy

Policy

Rare diseases – what are they?

Life-threatening or chronically debilitating diseases – mostly inherited – that affect so few people that combined efforts are needed to:

- reduce the number of people contracting the diseases
- prevent newborns and young children dying from them
- preserve sufferers' quality of life and socio-economic potential.

In EU countries, any disease affecting fewer than 5 people in 10 000 is considered rare. That number may seem small, but it translates into approximately 246 000 people throughout the EU's 27 member countries. Most patients suffer from even rarer diseases affecting 1 person in 100 000 or more.

It is estimated that today in the EU, 5-8000 distinct rare diseases affect 6-8% of the population – between 27 and 36 million people.

What is the EU doing?

Helping to pool scarce resources that are currently fragmented across individual EU countries. Joint action helps patients and professionals share expertise and information across borders. Specific measures include:

- improving recognition and visibility of rare diseases
- ensuring that rare diseases are adequately coded and traceable in all health information systems
- supporting national plans for rare diseases in EU member countries
- strengthening European-level cooperation and coordination
- creating European reference networks linking centres of expertise and professionals in different countries to share knowledge and identify where patients should go when expertise is unavailable in their home country
- encouraging more research into rare diseases
- evaluating current screening population practices
- supporting rare diseases registries and providing a European Platform for rare diseases registration.

Patient organisations are particularly important because they provide additional incentives for developing orphan drugs to combat rare diseases.

Legal basis of EU policy

- Communication on rare diseases: Europe's challenges COM(2008) 679 final
- Recommendation on an action in the field of rare diseases (2009/C 151/02)
- Other EU legal documents on rare diseases

e-newsletter 12 December 2013

ENIHR: Protecting citizens against new health r

Highlights

2013 Report on the State of the Art of Rare Diseases Activities in Europe - EUCERD Joint Action

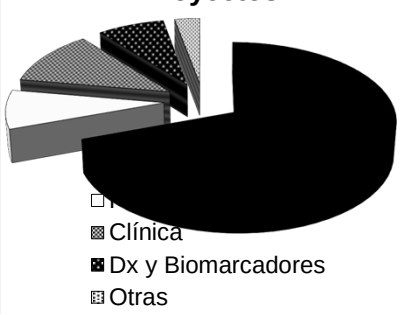
EUCERD Recommendations on core indicators for Rare Disease national plans/strategies

New web site of the International Rare Disease Research Consortium (IRDiRC)

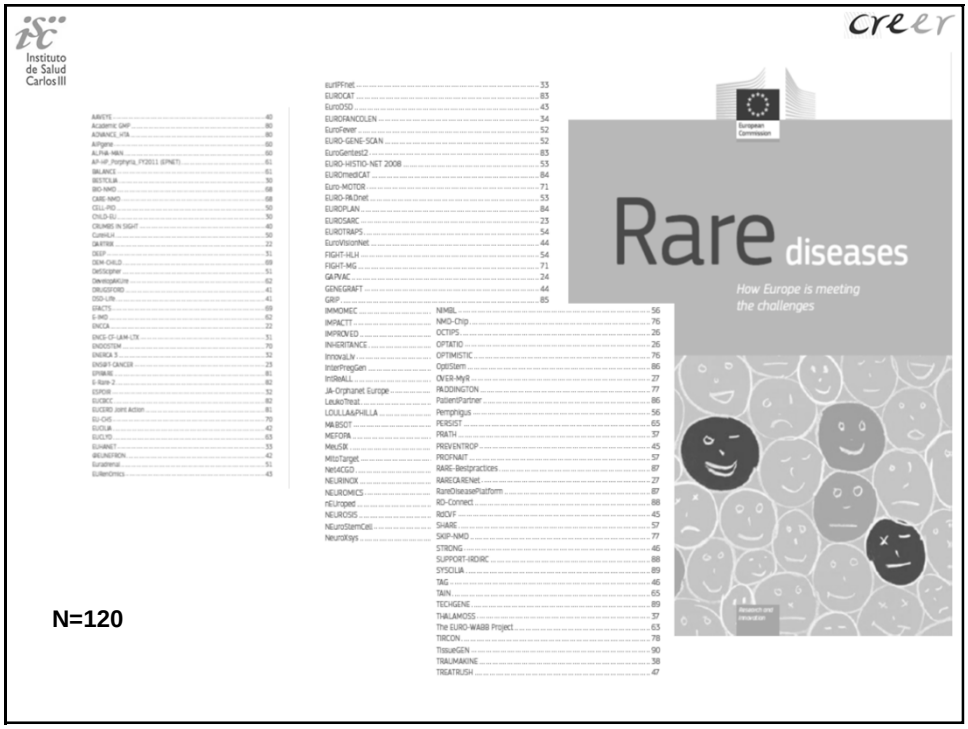
Related information

Press material | Key documents
Videos | Eurobarometers
Events | Consultations
Publications | Projects

EUCERD



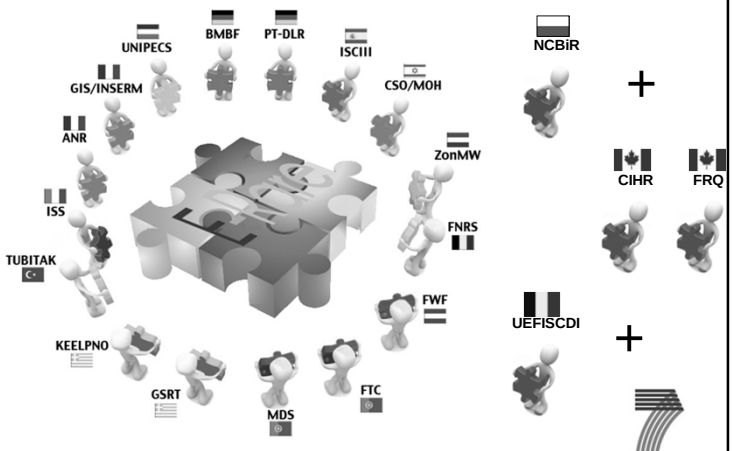
Basic research	Gene search	512
	Mutations search	595
	Gene expression profile	274
	Genotype-phenotype correlation	383
	In vitro functional study	1047
	Animal model creation/ study	492
	Human physiopathology study	733
Pre-clinical trial	Pre-clinical gene therapy	181
	Pre-clinical cell therapy	91
	Pre-clinical drug development	152
	Pre-clinical vaccine development	31
	Medical device/instrumentation development	25
Clinical research	Observational clinical study	448
	Epidemiological study	228
Diagnostics & biomarkers	Diagnostic tool / protocol development	301
	Biomarker development	149
Other	Health sociology study	80
	Health economics study	14
	Public health / health services study	75
	Total	4690



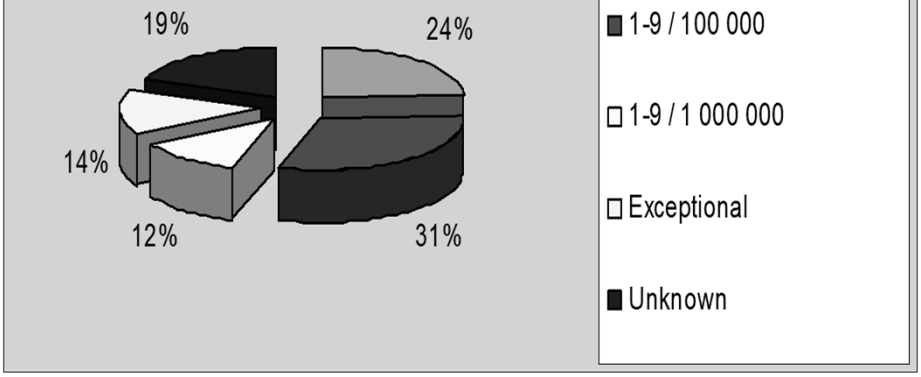
N=120

E-Rare-2
(Consorcio 2011 - 2014)

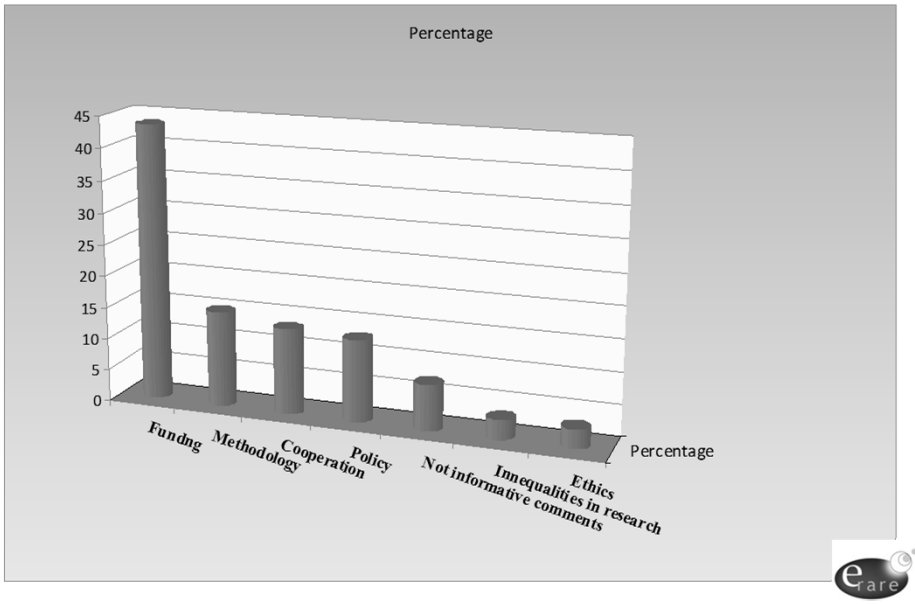
15 Estados Miembros UE y Países Asociados
20 Agencias Financiadoras Públicas / Ministerios



Proyectos según cifra de prevalencia de las Enfermedades



Tipo de Dificultades (Bottlenecks)




Public Health

EUROPLAN

EUROPLAN

European Project for Rare Diseases National Plans Development

Funded: European Commission Public Health Program 2003-08
Priority area: 1. Health Information (HI 2007).
Action: 1.4 Developing strategies for information exchange and responding to non-communicable health threats
Contract Number: 101120
Project Coordinator: Domenica Taruscio (ISS)
Main Partner: Istituto Superiore di Sanità (Italy)
Associated Partners: 10
Project Period: 1 April 2008 – 31 March 2011
Reporting Period: N.A.

Selecting indicators to evaluate the achievements of RD initiatives

EP-06-Indicators

Document Authors: Manuel Posada, Maria José Carrodeguas, Manuel Mas Pérez and EUROPLAN Working Group on Indicators
Revision Date: 22 February 2011
Diffusion Level: Public
Intended Target:
Key words: indicators
Relevance Task: T5.3 - Prepare a proposal of indicators to be discussed by experts

www.europlanproject.eu

EUROPLAN*: (2008-2011; 2012-2015) project, funded by the EU Commission*

57 Partners

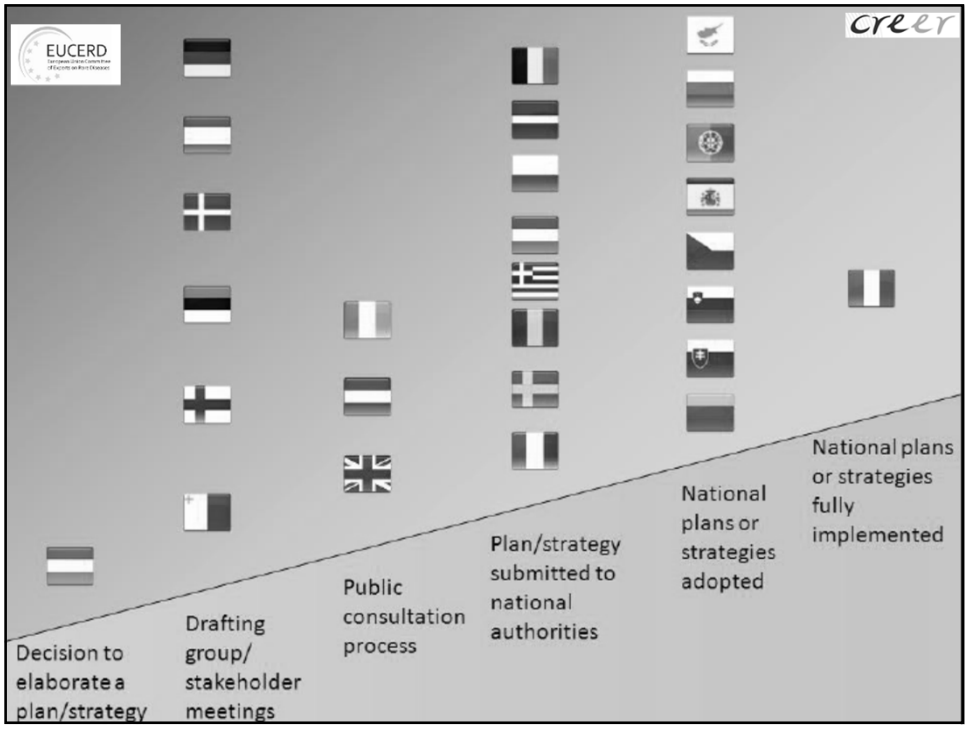
Associated & Collaborating:

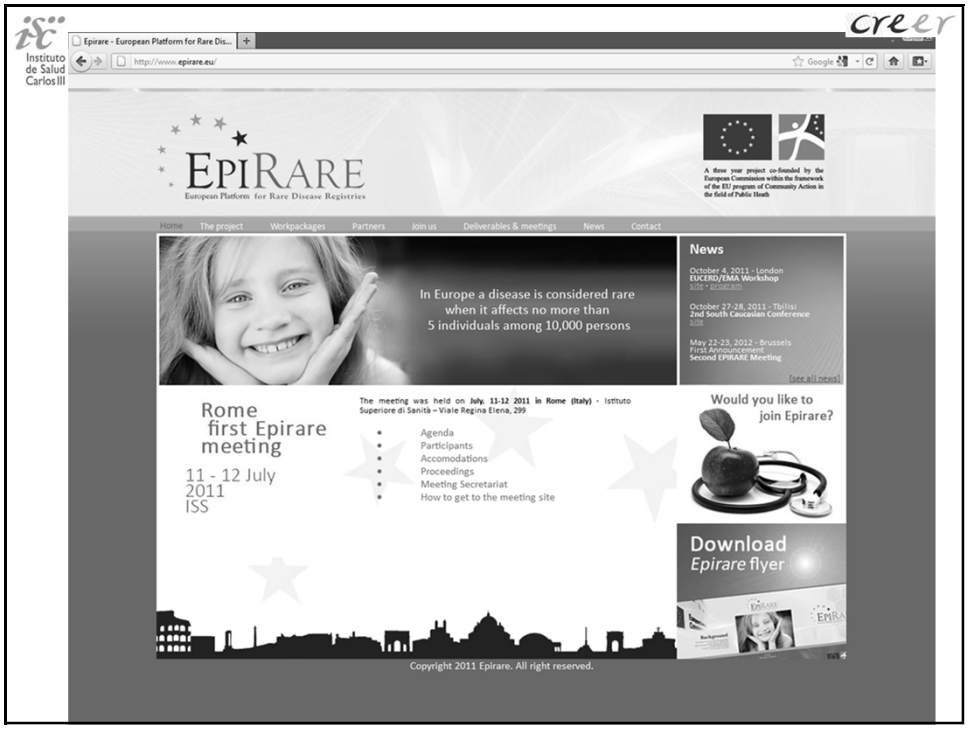
clinicians, scientist, health authorities, patients' Groups

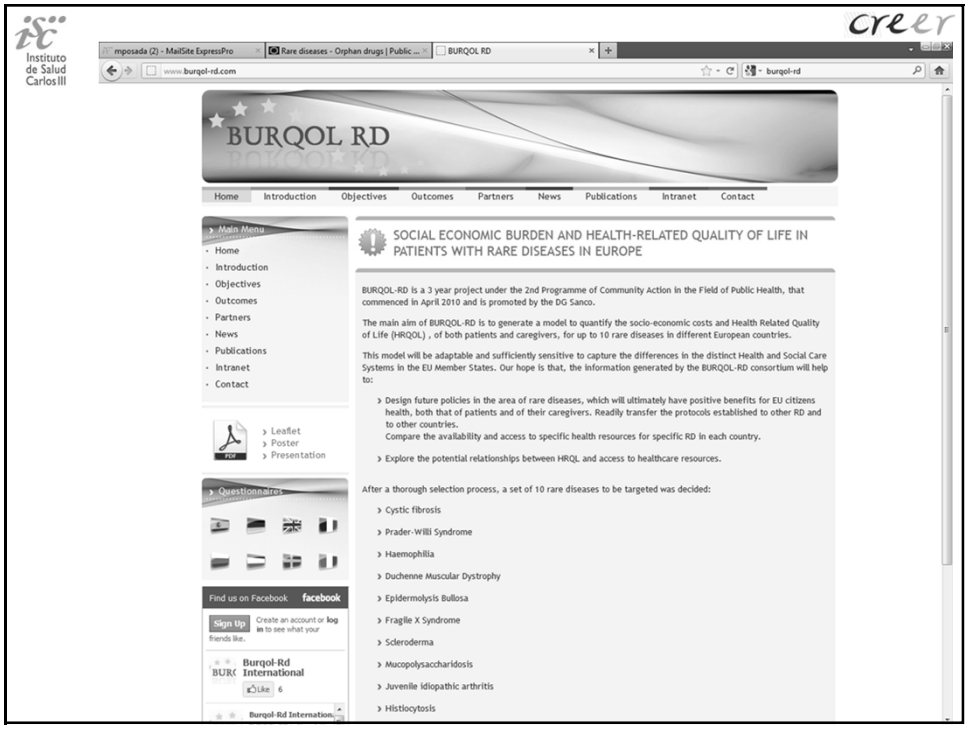
34 Countries

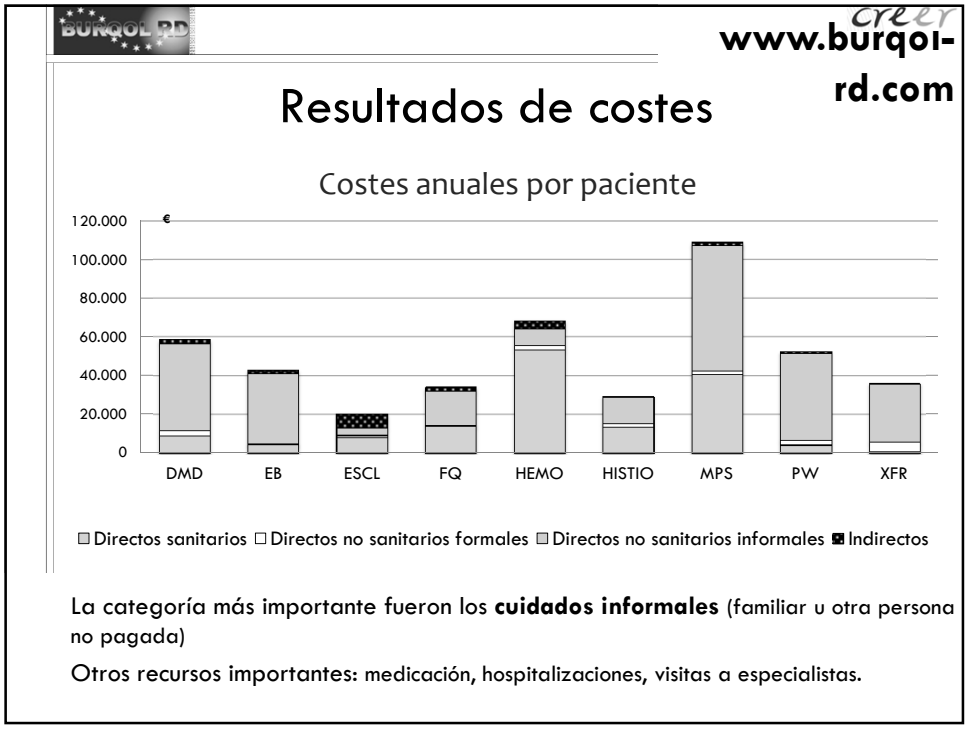
EURORDIS
Rare Diseases Europe

www.europlanproject.eu


EUCERD
creer
Decision to elaborate a plan/strategy
Drafting group/ stakeholder meetings
Public consultation process
Plan/strategy submitted to national authorities
National plans or strategies adopted
National plans or strategies fully implemented

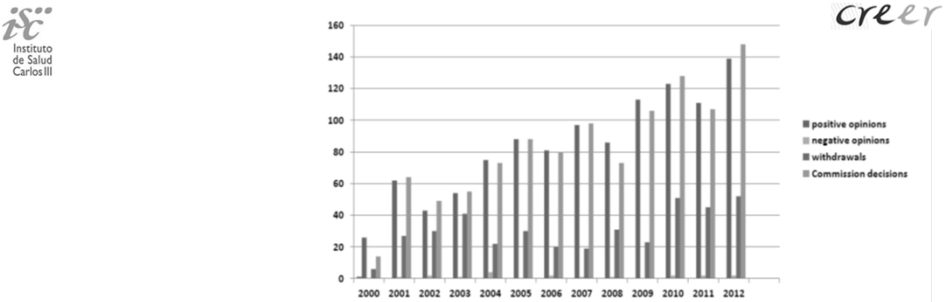

Instituto de Salud Carlos III
creer
Epirare - European Platform for Rare Dis...
http://www.epirare.eu/
EPIRARE
European Platform for Rare Disease Registries
A three year project co-founded by the European Commission within the framework of the EU program of Community Action in the field of Public Heath
Home
The project
Workpackages
Partners
Join us
Deliverables & meetings
News
Contact
In Europe a disease is considered rare when it affects no more than 5 individuals among 10,000 persons
News
October 4, 2011 - London
EUCERD/EMA Workshop
site - program
October 27-28, 2011 - Tbilisi
2nd South Caucasian Conference
site
May 22-23, 2012 - Brussels
First Announcement
Second EPIRARE Meeting
[see all news]
Rome first Epirare meeting
11 - 12 July 2011
ISS
The meeting was held on July, 11-12 2011 in Rome (Italy) - Istituto Superiore di Sanità – Viale Regina Elena, 299
Agenda
Participants
Accomodations
Proceedings
Meeting Secretariat
How to get to the meeting site
Would you like to join Epirare?
Download Epirare flyer
Copyright 2011 Epirare. All right reserved.

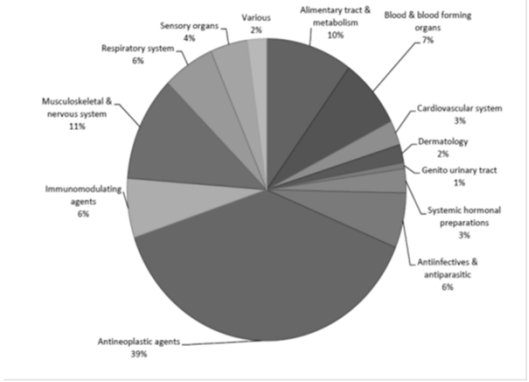




La categoría más importante fueron los **cuidados informales** (familiar u otra persona no pagada)

Otros recursos importantes: medicación, hospitalizaciones, visitas a especialistas.





Medicamentos Huérfanos

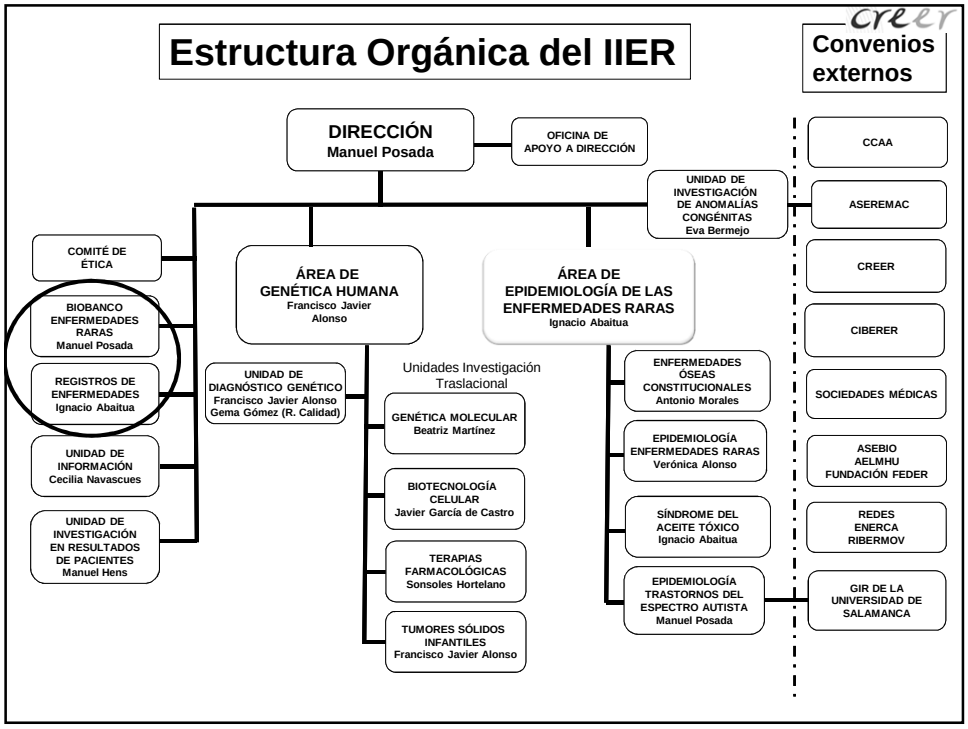
Acciones en España

Planes Nacionales y Autonómicos

- **Andalucía**
- **Extremadura**
- **Castilla-La Mancha**
- **Murcia**

- **Estrategia del Ministerio de Sanidad y Consumo (España)**

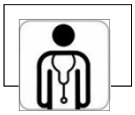
Estructura Orgánica del IIER



- Rare Diseases Epidemiology, 2010

Objetivo

Desarrollar el Registro Nacional de ER en España


Registro de Pacientes ⟹ **Registro de Base poblacional**

INVESTIGACIÓN

Historia natural de la enfermedad

Seguimiento

Ensayos Clínicos (reclutamiento)

Muestras biológicas

PLANIFICACIÓN Y POLÍTICA SANITARIA

Prevalencia

Incidencia

Mortalidad

Historia natural de la enfermedad

REGISTRO NACIONAL DE ENFERMEDADES RARAS
IIER - ISCIII

SpainRDR - Spanish Rare Diseases Registries Research Network – https://www.spainrdr.isciii.es
An initiative of the International Rare Diseases Research Consortium-IRDiRC

SpainRDR - Spanish Rare Diseases Registries Research Network – https://www.spainrdr.isciii.es
An initiative of the International Rare Diseases Research Consortium-IRDiRC

Biobanco Nacional de Enfermedades Raras

Red Europea de Biobancos de Enfermedades Raras – EUROBIOBANK

Red Nacional de Biobancos ISCIII (RetBioH)

Colección seriier
NIPO: 725-13-013-7
Depósito Legal: M-21011-2013

Instituto de Investigación de Enfermedades Raras
Avda. Monforte de Lemos, 5
28029 Madrid-España
biobanco_iier@isciii.es

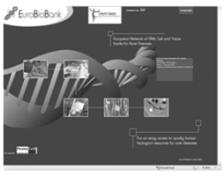
Instituto de Investigación de Enfermedades Raras

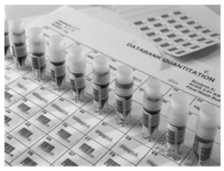
Biobanco Nacional de Enfermedades Raras

biobanco_iier@isciii.es

Participante en:
RD-Connect
SpainRDR
Red Biobancos
EuroBioBank

Red Biobancos
Instituto de Salud Carlos III
GOBIERNO DE ESPAÑA
MINISTERIO DE ECONOMÍA Y COMPETITIVIDAD



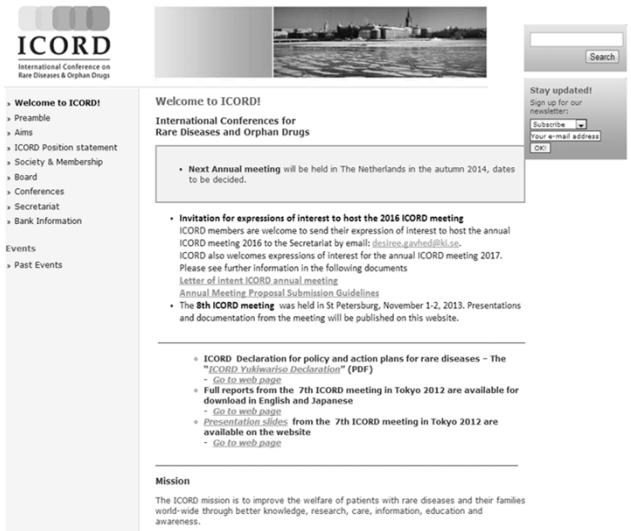


Presente y Futuro

Proyectos internacionales relacionados

SpainRDR - Spanish Rare Diseases Registries Research Network – https://www.spainrdr.isciii.es
An initiative of the International Rare Diseases Research Consortium-IRDiRC

ICORD (International Conferences for Rare Diseases and Orphan Drugs)



El International Rare Dieases Research Consortium [IRDiRC]

- Fue lanzado en Reikiavik, Islandia, 27-28 de Octubre 2010, convocada por NIH (USA) y EC.
- España fue el primer país en devenir miembro de pleno derecho en la siguiente reunión (Bethesda, USA, 6-8 de abril de 2011).

PAIS	Comité Ejecutivo, Miembros
EU	European Commission, DG Research and Innovation,
Australia	Western Australian Department of Health
Canadá	Canadian Institute of Genetics CIHR
Canadá	Genome Canada
EU	European Commission, DG Health and Consumer Protection
Europa	E-RARE-2 (ERA-Net group of funders alliance)
Francia	French National Research Agency [ANR]
Francia	French Association against Myopathies
Francia	Lysogene
Alemania	Federal Ministry of Education and Research [BMBF]
Irlanda	Shire
Italia	Telethon Foundation
España	Instituto de Salud Carlos III [ISCIII]
USA	Sandford Research
USA	Prosensa
USA	Food and Drug Administration [FDA]
USA	National Human Genome Research Institute, NIH
USA	National Cancer Institute, NIH
	Grupo de pacientes
Europa	EURORDIS
	Comités Científicos
	Chair Comité de Terapias
	Chair Comité Interdisciplinario
	Chair Comité de Diagnósticos
	Invited
	SUPPORT IRDiRC project

IRDiRC

Gobernanza

Comité Ejecutivo

- 1 representante por agente financiador (10 M $ en 5 años consecutivos)
- 1 representante por alianza de agentes financiadores (acumulado de 10 M $ en 5 años consecutivos)
- Los “chairs” de los 3 Comités Científicos

Commités Científicos

Diagnóstico, sequenciación y caracterización | **Interdisciplinario “Horizontal”** | **Terapéuctico**

15 miembros (representación equilibrada de científicos, pacientes, industria, etc.

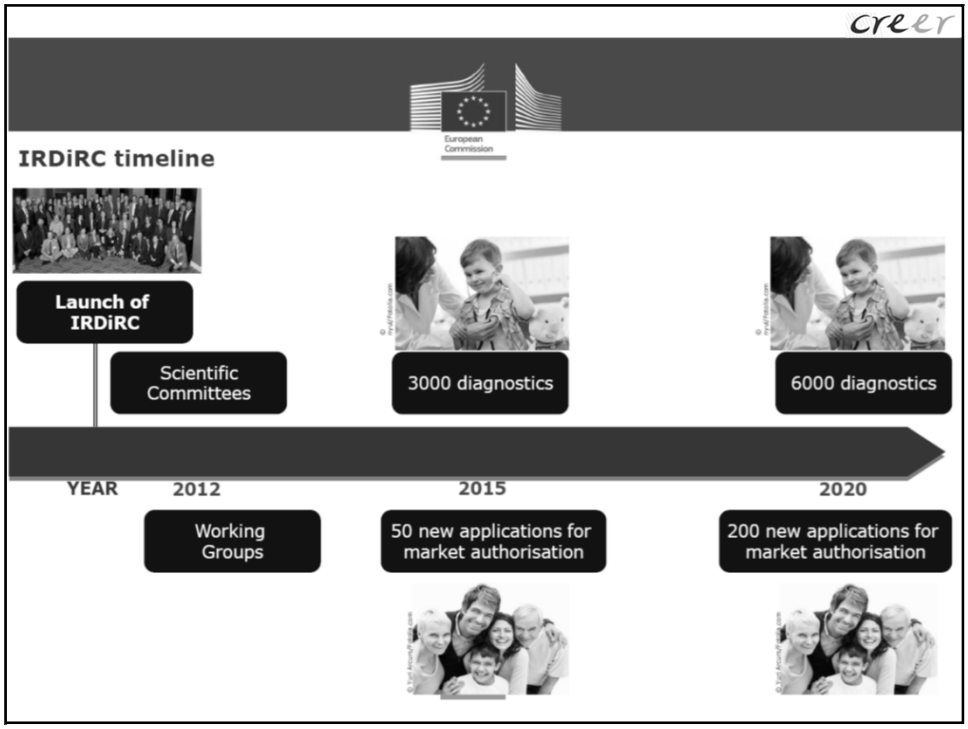
Grupos de trabajo

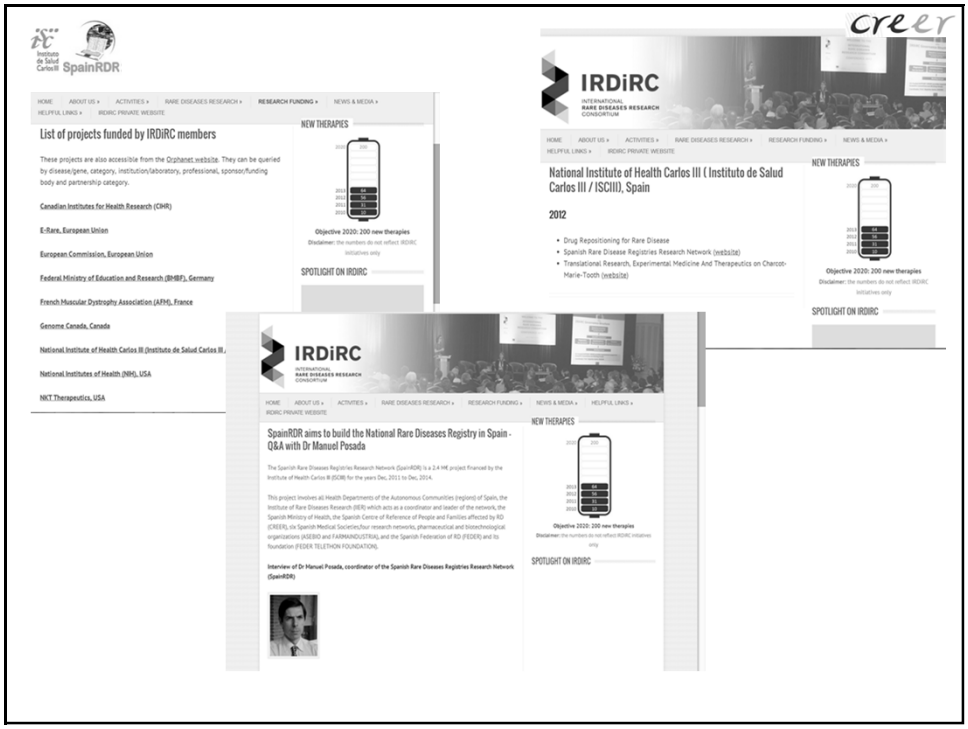
Ontologias | Genoma | Sistemas modelo | Clinico

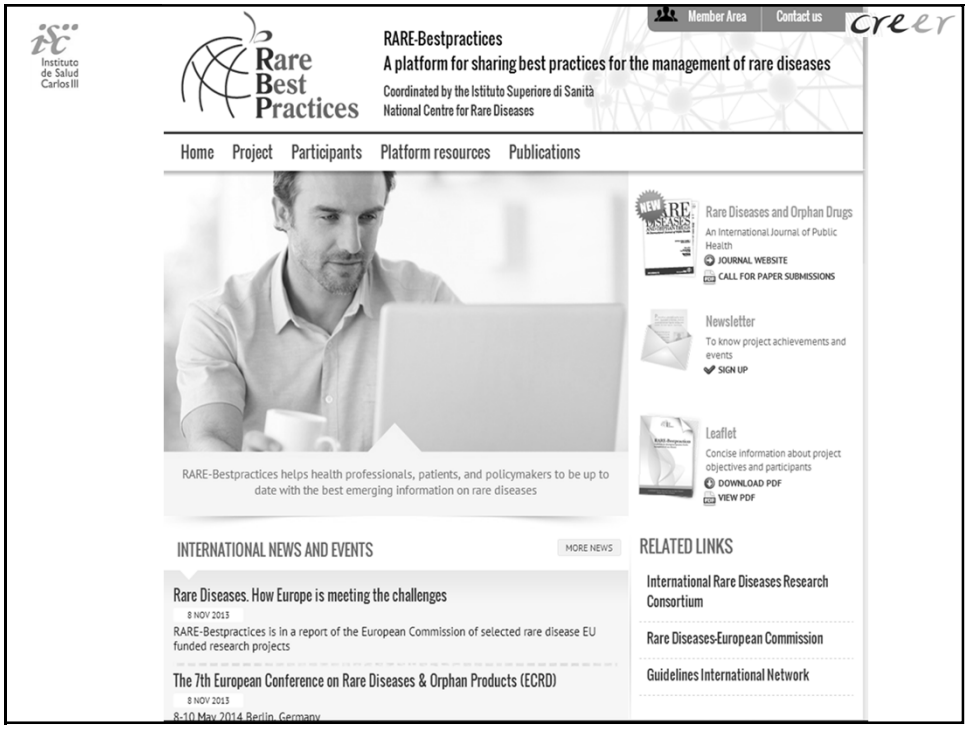
Registros | Secuenciación | Biomarcadores | Terapias

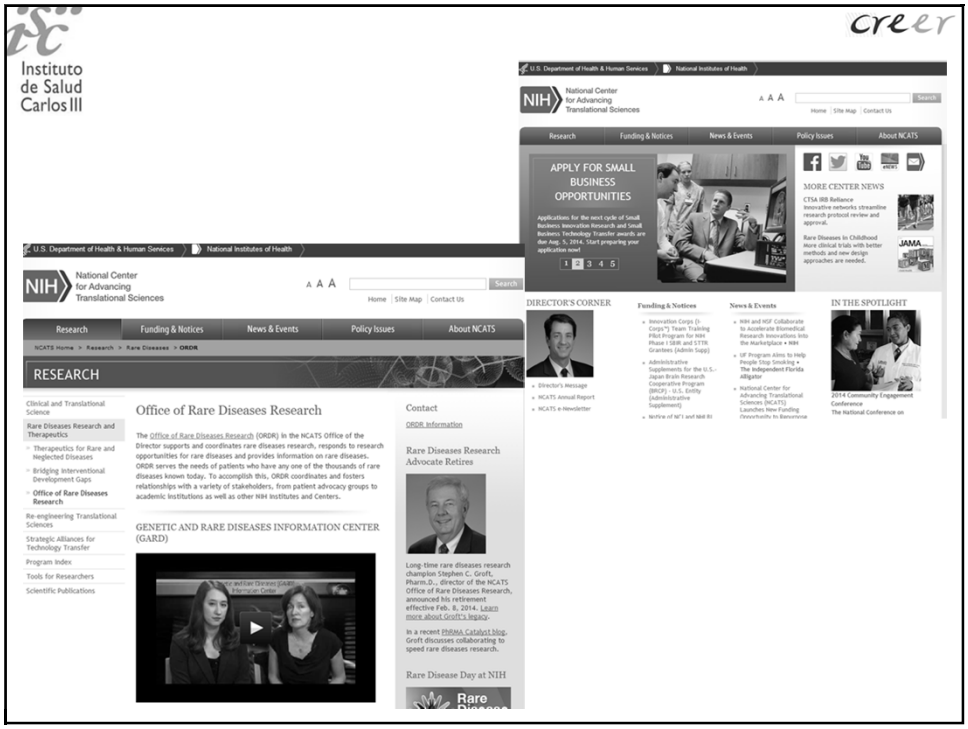
Etica | Biobancos | Bioinformática | Regulatorio

Represensantes de proyectos financiados


creer
European Commission
IRDiRC timeline
Launch of IRDiRC
Scientific Committees
3000 diagnostics
6000 diagnostics
YEAR
2012
2015
2020
Working Groups
50 new applications for market authorisation
200 new applications for market authorisation


SpainRDR
List of projects funded by IRDiRC members
Canadian Institutes for Health Research (CIHR)
E-Rare, European Union
European Commission, European Union
Federal Ministry of Education and Research (BMBF), Germany
French Muscular Dystrophy Association (AFM), France
Genome Canada, Canada
National Institute of Health Carlos III (Instituto de Salud Carlos III /
National Institutes of Health (NIH), USA
NKT Therapeutics, USA
IRDiRC
National Institute of Health Carlos III (Instituto de Salud Carlos III / ISCIII), Spain
2012
Drug Repositioning for Rare Disease
Spanish Rare Disease Registries Research Network (website)
Translational Research, Experimental Medicine And Therapeutics on Charcot-Marie-Tooth (website)
NEW THERAPIES
SPOTLIGHT ON IRDiRC
SpainRDR aims to build the National Rare Diseases Registry in Spain - Q&A with Dr Manuel Posada


Instituto de Salud Carlos III
Rare Best Practices
Member Area
Contact us
creer
RARE-Bestpractices
A platform for sharing best practices for the management of rare diseases
Coordinated by the Istituto Superiore di Sanità
National Centre for Rare Diseases
Home
Project
Participants
Platform resources
Publications
RARE-Bestpractices helps health professionals, patients, and policymakers to be up to date with the best emerging information on rare diseases
Rare Diseases and Orphan Drugs
An International Journal of Public Health
JOURNAL WEBSITE
CALL FOR PAPER SUBMISSIONS
Newsletter
To know project achievements and events
SIGN UP
Leaflet
Concise information about project objectives and participants
DOWNLOAD PDF
VIEW PDF
INTERNATIONAL NEWS AND EVENTS
MORE NEWS
Rare Diseases. How Europe is meeting the challenges
8 NOV 2013
RARE-Bestpractices is in a report of the European Commission of selected rare disease EU funded research projects
The 7th European Conference on Rare Diseases & Orphan Products (ECRD)
8 NOV 2013
8-10 May 2014 Berlin, Germany
RELATED LINKS
International Rare Diseases Research Consortium
Rare Diseases-European Commission
Guidelines International Network


Instituto de Salud Carlos III
creer
U.S. Department of Health & Human Services
National Institutes of Health
NIH
National Center for Advancing Translational Sciences
Research
Funding & Notices
News & Events
Policy Issues
About NCATS
APPLY FOR SMALL BUSINESS OPPORTUNITIES
MORE CENTER NEWS
DIRECTOR'S CORNER
IN THE SPOTLIGHT
RESEARCH
Office of Rare Diseases Research
GENETIC AND RARE DISEASES INFORMATION CENTER (GARD)
Contact
Rare Diseases Research Advocate Retires
Rare Disease Day at NIH

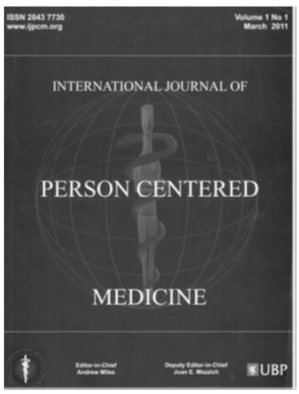
Prioridades para la investigación en enfermedades raras2014-2020

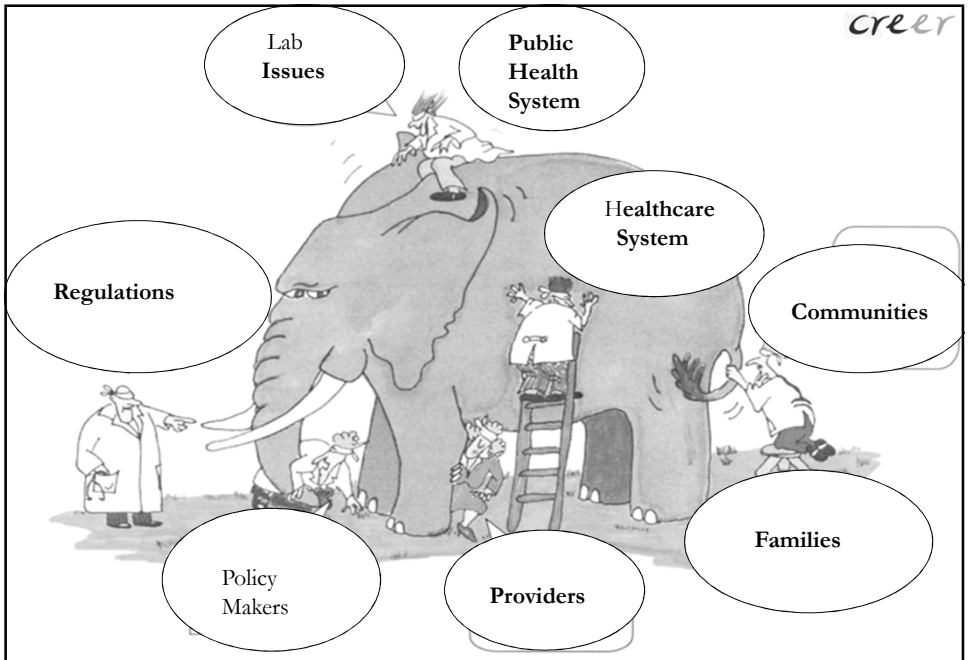
- Apoyo a los registros y otras infraestructuras
- Aumentar el conocimiento de los mecanismos de las enfermedades raras Trasladar los resultados de investigación en tratamientos para los pacientes
- Diseñar estrategias amplias de ensayos clínicos
- Investigación en ciencias sociales

Cómo conducir la investigación en enfermedades raras

- Empoderando a los pacientes en investigación
- Integrando acciones
- Sosteniendo a largo plazo infraestructuras y proyectos
- Intrumentos de financiación

Medicina Centrada en la Persona





Implementation: Blind Men and the Elephant

http://www.nature.com/ki/journal/v62/n5/fig_tab/4493262f1.html

Unidos dejamos huella

FEDER

Muchas Gracias

Institute of Rare Diseases Research (IIER)

Instituto de Salud Carlos III

Manuel Posada

mposada@isciii.es