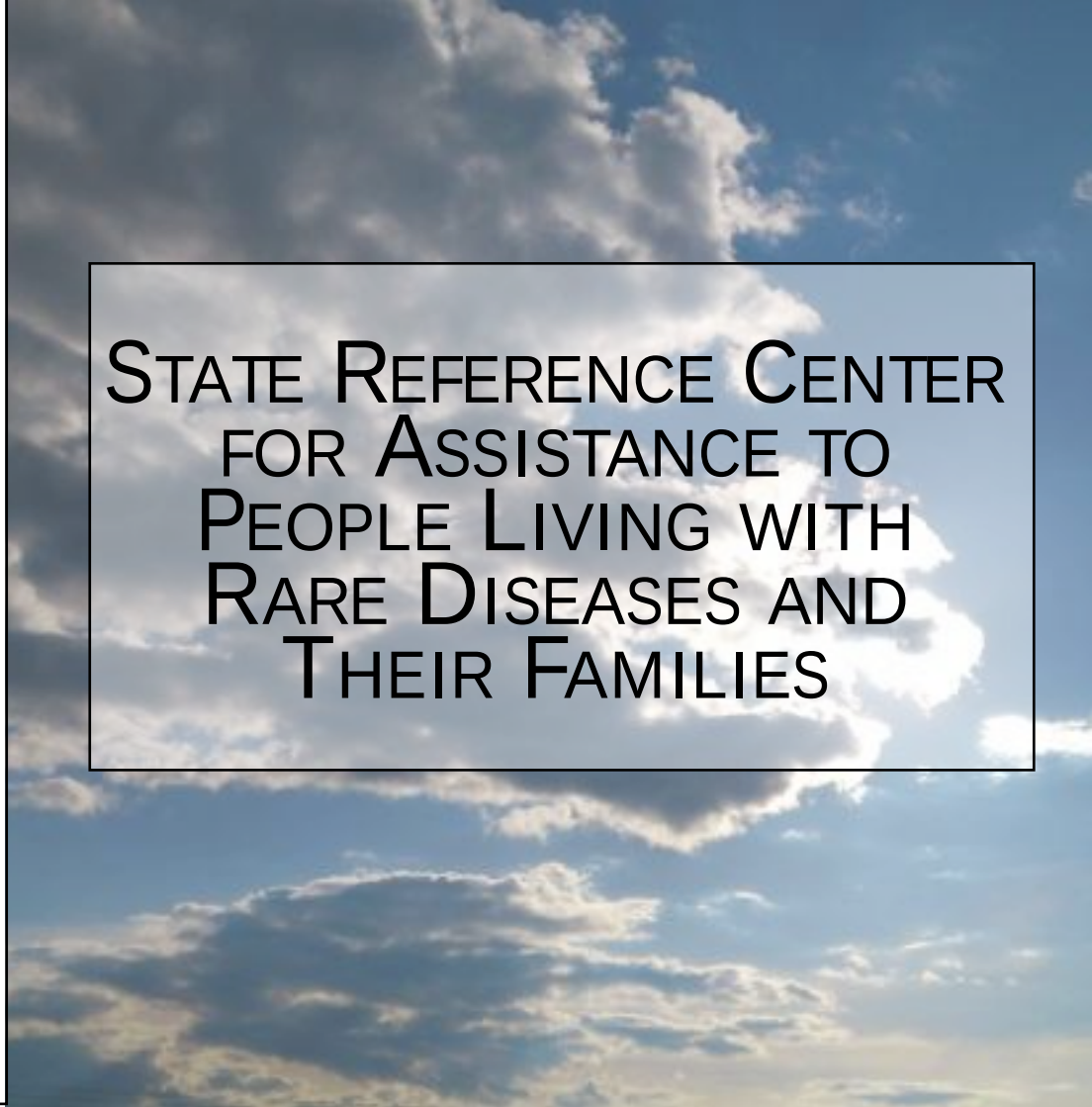

CENTRO DE REFERENCIA ESTATAL DE ATENCIÓN A PERSONAS CON ENFERMEDADES RARAS Y SUS FAMILIAS

STATE REFERENCE CENTER FOR ASSISTANCE TO PEOPLE LIVING WITH RARE DISEASES AND THEIR FAMILIES

Report
Actions
2013

2013
AÑO ESPAÑOL
DE LAS ENFERMEDADES RARAS

GOBIERNO DE ESPAÑA
MINISTERIO DE SANIDAD, SERVICIOS SOCIALES E IGUALDAD
SECRETARÍA DE ESTADO DE SERVICIOS SOCIALES E IGUALDAD
IMSERSO

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Presentation

Aitor Aparicio | Director

The State Reference Center for Assistance to People Living with Rare Diseases and Their Families is a public, state-owned service, depending on the IMSERSO (Social Services and older Institute), focused on improving the quality of life of people living with rare diseases , their families and professionals who serve them.

As a State Reference Center it is constantly searching new solutions that make life easier for those who support these rare diseases. Some of the tasks are: performing tasks of coordination and research, innovation, support of professional training, knowledge promotion and develops awareness activities, providing technical support to other entities.

As a Specialized Center in the care of people with rare diseases and their families, develops different programs of care and support to families, carers and people affected by rare diseases from a social-medical approach.

Our priority is a Comprehensive Care focused on Person (AICP) work with a model focused on innovation, quality and participation.

In the year 2013, which was declared by the Minister of Health, Social Services and Equality, Spanish Year Rare Disease, we highlight the high participation and involvement they have had in our Center Associations, Foundations and other entities representing interests of people with rare disease, and who have contributed to give life to CREER.

We believe that reciprocity and networking is the most efficient way to find solutions to the complex difficulties that people face affected with a rare disease.

Therefore, we are determined to encourage the participation of individuals and entities in developing actions CREER, to cooperate with others and work to create synergies, to improve the quality of life for people with rare diseases and their families.

With this document we want to present, in summary, the actions taken during the year 2013.

Focused on people, committed to improve quality

Organization chart of the Creer

Manager
Director

Management
Finance

Technical Area I
Coordinator Area
Social Educator

Social and Health Area
Coordinator Area
Physical Therapy Doctor
General Practitioner
Doctor
Psychologist
Social Worker
Speech Therapist
Physiotherapists
Occupational Therapists
Pedagogue
Teachers
Nurses
Nursing Assistants

Technical Area II
Coordinator Area
Documentary

They have actively participated in the design, implementation and evaluation of actions
People Living with rare disease, associations represent their interests and their families
Professionals
Volunteers
Universities
Public Administrations
and Other non-profit Organisations.

CREER has developed an Integrated Management System based on the rules UNE-EN-ISO 9001, UNE-EN-ISO 14001, UNE 170001

Focused on people, committed to improve quality

3094
People assisted

4102
Participant people in awareness and spreading actions

Information and orientation
Meetings
Courses and workshops
Therapy

486
Professionals

Cooperation
Information
Training

45
Associations

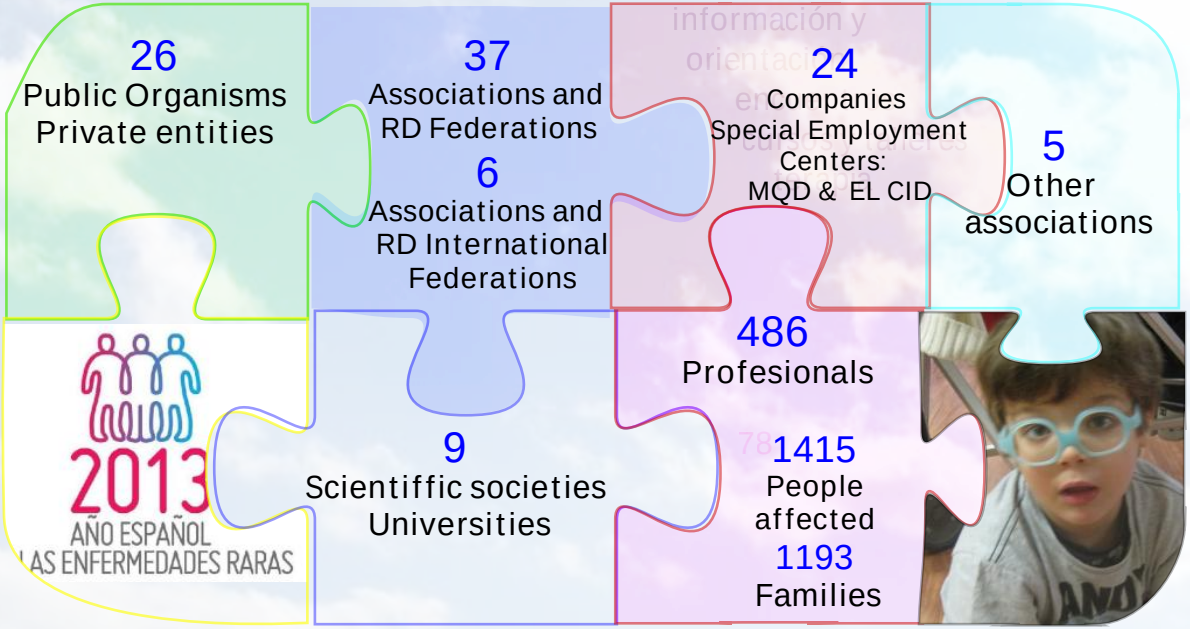
Participating entities

Fundación Síndrome de Dravet
Asociación Española Extrofia Vesical. ASEXVE
Asociación Ataxia Galicia. AGA
Asociación Española Síndrome de Prader Willi. AESPW
Asociación Enfermedades Raras y Otros Trastornos Graves del Desarrollo. DGENES
Asociación Española Síndrome de Stickler.
Asociación Nacional de Arnold Chirari. ANAC
Plataforma Afectados de ELA
Asociación Española de Familiares y Enfermos de Wilson. AEFEW
Asociación Española de Linfangioleiomiomatosis. AELAM
Asociación Española Síndrome Defectos congénitos de la glicosilación AESCDG
Asociación Enfermedad de Huntington de CyL. E.H. CyL
Huntington´s Disease Youth Organization.
Fundación Síndrome de West. FSW
Asociación Española de Esclerodermia. AEE.
Asociación Española Síndrome de Wolf-Hirschorn. AESWH
Asociación Asturiana Arnold Chiari. CHySPA
Asociación DEDINES
Asociación española Paraparesia Espástica Familiar. AEPEF
Asociación Duchenne Parent Project España
Asociación Española Leucosdistrofia. ELA ESPAÑA.
Federación Española de Enfermedades Raras. FEDER
Federación Española de Ataxia. FEDAES
Asociación Síndrome de Costello y Cardio Facio Cutaneo ACyCFC
Asociación Española Síndrome de Sotos. AESS
Asociación Española Síndrome de Smith Magenis. ASME
Asociación Cantabria Síndrome de Williams. ASW-Cantabria
Asociación Española Síndrome de Williams. AESW
Asociación Española Patologías Mitocondriales. AEPMI
Fundación Ana Carolina Díez-Mahou
Asociación Española Déficits Inmunitarios Primarios. AEDIP
Asociación Española Sindrome de Phelan Mc Dermid.
Asociación Española de Pénfigo Penfigoide y Otras enfermedades vesiculoampollosas. AEPPEVA
Asociación Nevus Gigante. ASONEVUS.
Asociación Paraparesia Espástica Familiar CyL. APEFCyL.
Asociación Española de Beneficencia Guatemala. AEB
IMSERSO CCAA EQUIPOS DE VALORACIÓN DE LA DISCAPACIDAD
Ministerio de Sanidad SS e Igualdad
Gerencia de Salud. CyL
European Rare Diseases Organisation EURORDIS
Hospital Universitario de Burgos
Instituto de Investigación en Enfermedades Raras Carlos III. IIER.
Sociedad Española de Neurología. SEN
Asociación Española de Neurogenética. NEUROGENES
Centros de Investigación Biomédica en Enfermedades Raras. CIBERER
Centros de Investigación Biomédica en Red Enfermedades Neurodegenerativas. CIBERNED
ASPANIAS Burgos
Federación Autismo CyL
Asociación de Autismo
Asociación de Daño Cerebral
Asociación Amigos del Camino de Santiago.
Universidad de Burgos. UBU
Universidad a Distancia. UNED
Universidad de Salamanca

Report Actions 2013

Focused on people, committed to improve quality

They have made it possible ...



Report Actions 2013

Developed actions

Meetings Programs

Within family Care Services, NGOs and people with rare diseases, CREER, in coordination with different associations and organisations representing people affected by rare diseases, during the year 2013 has conducted:

- Thirty-two Meetings of National Rare Disease Associations.
- International Medical Meeting, Hispanic Network of Pathologies Rare Diseases. Guatemala Foundation Rare Diseases
- International Meetingl. Huntington´s Disease Youth Organization
- IX National Congress Medical. Arnold Chiari National Association. ANAC
- Scientific medical Conference on Mitochondryal Pathologyes. Spanish Association Mitochondryal Pathologyes. AEPMI
- Scientific medical Conference on ALE. Platform ALE
- III Congress of Glycogen Storage Diseases. Spanish Association of GSD
- Medical Meeting, Researchers and Families. Spanish Association of Ataxia telangiectasia Family. AEFAT
- Scientific Medical Conference on Sotos Syndrom. Spanish Association Sotos Syndrom.
- Scientific medical Conference on Linfangioleiomiomatosis. Spanish Association Linfangioleiomiomatosis. AELAM
- MusicTherapy Conference in Williams Syndrome. ASW-Cantabria and AESW
- Scientific medical Conference on Wolf-Hirschhorn Syndrom. Spanish Association Wolf-Hirschhorn Syndrom AESWH.

People affected	628
Families	915
Professionals	233
Total	1776

Developed actions

NacER Program

This program is a colaboration with the Neonatology Unit Care Complex University of Burgos (CAUB) serving infants with real or suspected diagnosis of a rare disease from locations near Burgos: Aranda de Duero, Miranda de Ebro, Soria, Palencia, Leon. While babies remain admitted to hospital for prolongued periods of time, CREER offers accommodation to mothers and fathers, and psychological service, guidance and counseling service, available resources (contact with other families, associations or support groups, ...), special care training required to meet the newborn (rehabilitation, early speech care therapy, ...).

In CREER were treated a total of 39 people, families from 12 babies admitted to the University Hospital of Burgos

Developed actions

SAMER Program

Multidisciplinary Care Service Rare Diseases (SAMER), aimed for affected people and family, has offered groups or individual psychological care, speech therapy, educational assistance, physiotherapy, occupational therapy, psychomotor, and multisensorial stimulation.

The program has responded to demands of people all over the national territory, to anyone who has required so providing accommodation in CREER.

Attended people
291

Physiotherapy	1250
Psychological therapy	645
Occupational therapy	731
Adaptive Learning Program	484
Speech therapy	465
Medical consultant	361
Total sessions	3936

Developed actions

Family Respite Care Program

During 2013 a total of 80 people have benefited from Family Respite Care Program that has been developed in the months of July and September.
The program aims to improve the quality of life of the people affected by a rare disease as their family and carers

Four shifts:
1 - 20 affected and five relatives
2 - 20 people affected
3 - 19 people affected
4 - 14 people affected and 7 relatives

Developed actions

Self-Care Program and Quality of Life

It provides training to carers and people with rare disease for:

- Help carers by providing information and resources that will serve them in their duty
- Help affected people by providing information that helps them to improve their welfare and independence.

The program is delivered over 30 hours and is divided into various multidisciplinary workshops. 3 training editions have been developed for four days between June and November 2013.

Workshops included in the program

- Transfer aids, safe patient handling and movement.
- How physiotherapy can help me in everyday
- Healthy lifestyle habits
- Time management
- A sense of humor as selfcare
- Social Resources
- Social networks management
- Multisensory stimulation
- Stress Management and Emotional support and adaptation processes to disease.
- Body Language and Psychomotricity
- Techniques to develop creativity
- Adaptive learning program

Have participated 24 affected individuals and 28 families.
Age Group: 3 to 12 years old, 13 to 24 years old and from 25 years

Developed actions

Training Actions

Fourth Edition of the Training School CREER-FEDER: "Training People, Inspiring Actions"
19th and 22th September
The professional and representatives 50 associations, who atended most state level, represented nearly 20,000 partners and families affected with rare diseases.

Contents: analysis and knowledge of legislation on themes related to cross Border Assistance, Data Protection, Disability Assessment and proposals for improvement.
An overview of the Action Plans, Key Messages and priorities was made as inspiration for Europlan2014 in Spain to realize. Impact of the 13 proposals to improve the situation of families with RD made with the consensus of more than 230 associations, coinciding with the declaration of 2013 as the Spanish Year of Rare Diseases. Project results as BURQOL-RD, on the economic impact and the quality of life of patients and families with rare diseases were spread and share. professional and families

People affected and families	64
Professionals	41
Total	105

Developed actions

Training Actions

Rare Disease training and disability

During September to December, in coordination with the Training Section of Imserso, there have been three editions of the Course "Rare Diseases and Disabilities. Procedure disability assesment in rare diseases"
The target group existed of professionals Assessment Disability Teams of the Autonomous Communities.

Held on 24 and 25 September 22 and 23 October and 19 and 20 November, 2013.

The main objetive of the course was essentially the gathering of technical criteria for assessing the situation of disability and dependency for discussion and reflection on professional evaluators in order to achieve better coordination in evaluation procedures.

Participants 110

Developed actions

Training Actions

II EDITION COURSE NEUROGENETICS

From 14th to 17 th June. With the colaboration of Neurogenetics Group of SEN, NEUROGENES, CIBERNED, CIBERER and CREER.
Goal: To help neurologists get familiar with the terminology and basic concepts, are handled in human genetic studies. Help them understand the genetic terminology and bring that language of molecular biology laboratory to the clinic and to the research and understandable way, with examples based on real cases taken from real hospitals.

Participants 51

Developed actions

Training Actions

Aimed at Professionals of the CREER

Communication Course in Sign Language
Team Management Course
Best s Practices Course

Participants:
30 professionals of the Creer

Developed actions

Training Actions

OTER TRAINING ACTIVITIES

In the frame of the colaboration with local authorities, the Centre provides rooms to Burgos Associations, to develop training activities:

- Multiple Sclerosis Awareness World Day.Múltiple Sclerosis Association Burgos. AFAEM
- Training courses for Autism Professional Association Burgos
- Meeting "Thinking Health Feeling people". Huntington Disease Association Castilla y León.
- Workshop phisotheraphy. AIDS Sheltered Housing "La Encina"
- Meeting "Hemophilia Information". Hemophilia Disease Association Burgos. HEMOBUR

Developed actions

Awareness and Dissemination Program

2013 Spanish Year or Rare Disease. Awareness activities with national and international reach which aims to bring the reality of the rare diseases to the general population

World Day of Rare Diseases:

- I National Meeting of families and affected . World Day RD. From 24th to 28th February.
- Radio programs such as "Today is not any day "of RNE. Saturday, 27th February
- Open House Day
- World Day of Rare Diseases Gathering Burgos. Lecture: Jordi Aubeso national and international athlete.
- EXHIBITION "Rare Diseases!?

From 20 February to 30 March at the University of Burgos.

From July 16 to August 15 in the Exhibition Hall of the Cathedral of Burgos.

Total Views: 3,153 People from 41 countries.

Report Actions 2013

Developed actions

Awareness and Dissemination Program

• Tour Camino de Santiago: "A Walk in the Rare Diseases". July 21th. Santiago Way in the town of Burgos led by the Association Camino de Santiago Burgos. The day was completed with children's activities, musical performances and theater.
Vídeo: http://www.youtube.com/watch?v=OtZO8_VWbHQ

• Awareness and information conferences Burgos and province.

• Video forum during the months of October and November. Movies: "Untouchable","Life Wings" and "Soul". Two films and one documentary showing different aspects surrounding rare diseases. Total participants: 169

• Present in: national and local mass media, television, newspapers, radios

• Presence on Social Networks

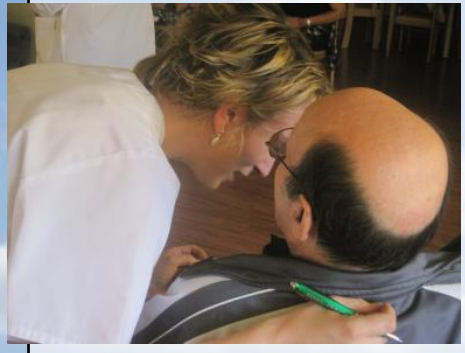
• CREER "Newsletter" 12 issues

• Schools Awareness Activities Rare Diseases. Participants 463

• Educational visits to the Center under the Awareness and dissemination Program, aimed for students, professionals from the health system, other professionals, teachers, etc.. There were 10 tours for a total of 160 participants.

Information and Advisory Service SIA

This service provides information, guidance and advice. CREER has responded to demands such as:
- People affected by rarely diagnosed disease
- Families of people affected by rare disease.



- Professionals (associations, public administration...)
- Other people (students, population, in general, ...).

Demands met
- People affected: 128
- Families: 204
- Professionals: 51
- Others: 17

Atentions
- Face to face: 80
- e-mail: 112
- Phone: 210

Total
- In the year 2013 have been treated a total of 400 demands

Cooperation with Other Entities

ER BIOBANK and Registration

This program is a colaboration with the Research Institute in Rare Diseases Carlos III (IIER) . Affected people, family who visit the center are offered information about the existence of the RD Register and what it is (get as much information as possible for further research, participate in studies ...)

They are provided with forms to complete and CREER is responsible for sending them to IIER .

CREER also offer the possibility to collect blood samples to be sent to the biobank IIER that belong to the First operational network in Europe. It also provides with human DNA cells and tissue samples, as a service to the scientific community for the development and research of rare diseases. It is a European Network dedicated exclusively to research on Rare Diseases in Europe.

Over 2013 were collected a total of 71 samples
124 cases were reported to the RD IIER

Cooperation with Other Entities

EURORDIS - Training Workshop WJA6. Copenhague

EURORDIS Training workshop about Social Service Providers during the 10th and 11th of October in Copenhagen developed.
This Working Group is one of the 8 working groups formed from the Joint Action Plan Working for Rare Diseases (2012-2015) promoted by the European Committee of Experts on
Rare Diseases (EUCERD). Committee was formally established by Commission Decision European of 30 November 2009 (2009/872 / EC) in order to assist the Commission in the preparation and implementation of Community activities in the field of rare diseases, in consultation and cooperation with the specialized agencies of the Member States, the competent European authorities in the field of research and public health measures and other stakeholders active in the field.

Link to the final document "Training for Social Services Providers Report"

http://www.eucerd.eu/wp-content/uploads/2014/03/WP6TrainingSSP.pdf

Cooperation with Other Entities

Participation Initiatives

• Experience Map Units. Its aim is to facilitate access to information, to know where there is focus of cases affected by these diseases and where there is a greater experience in the diagnosis and treatment. Data are available of all the Spanish Regions to eight groups of diseases (ataxia, Angelman syndrome, muscular dystrophies, myasthenias, muscle atrophy, myotonia and fragile X Syndrome).

• Updated National Strategy on Rare Diseases of the National Health System. The strategy, part of the NHS Quality Plan, is a coordination tool for the NHS. It was approved in 2009 by the Inter-Territorial Council of the National Health System (CISNS), the result of consensus between all stakeholders: Ministry of Health and Social Policy, Regional Governments, scientific societies, patient organizations and representatives of other ministries.

According to the monitoring and evaluation results, the Creer has colaborated on updating both the content and the possible improvement actions of the National Strategy.

Contact

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con Enfermedades Raras y sus Familias

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