



asociación española
enfermos **glucogenosis**

Asociación Española de Enfermos de Glucogenosis (AEEG)

Spanish Association of GSD affected

Alberto Molares, PhD

(Scientific Program Coordinator)

Very brief history of AEEG

- Born in May 1999
- Headquarters: Alcantarilla (Murcia)

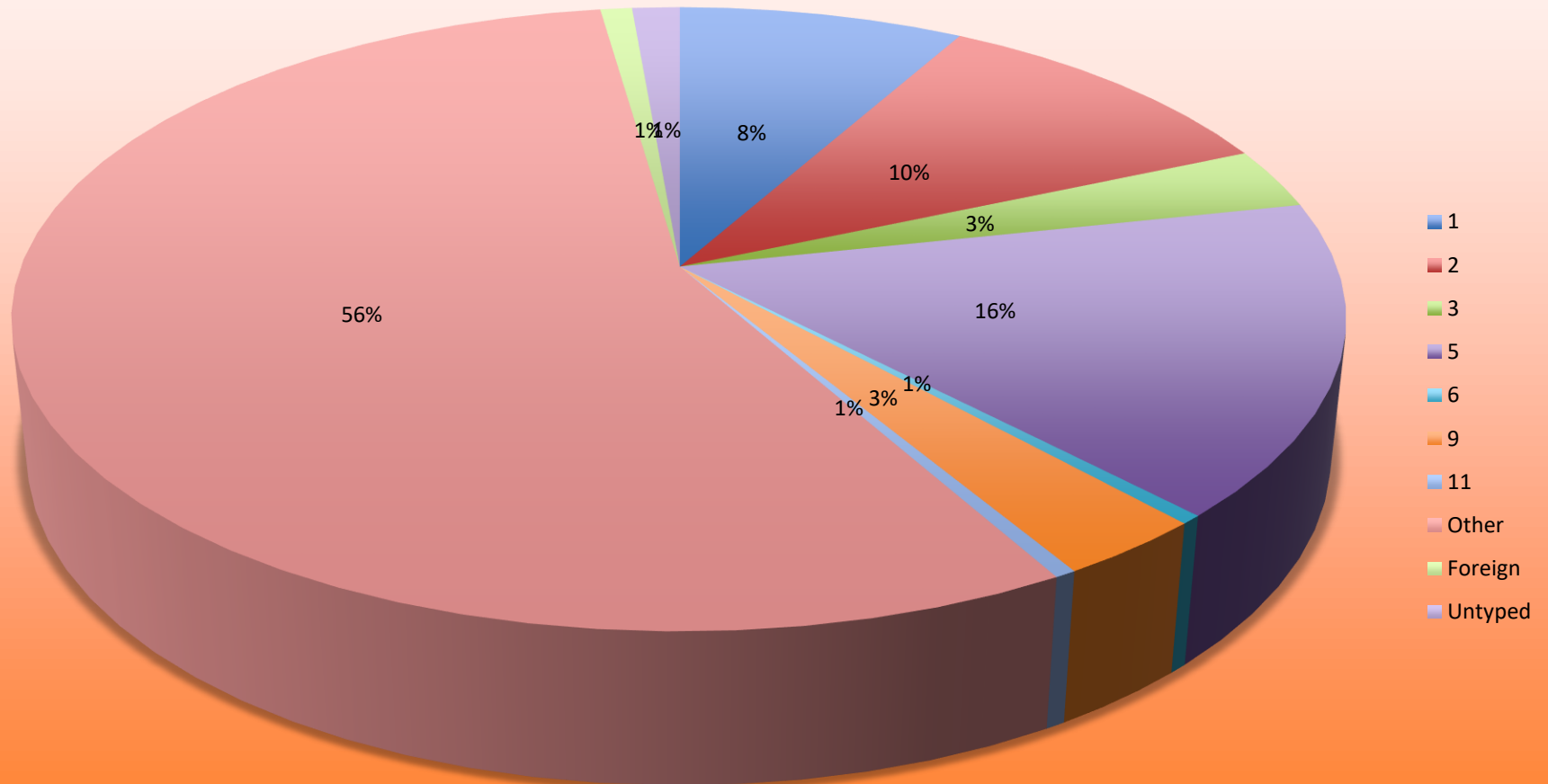


Relevant data

- 249 associates
- 42% GSD affected
- 1% Foreign GSD affected (Latin-America)
- 1% mistyping GSD affected
- 56% family or GSD related

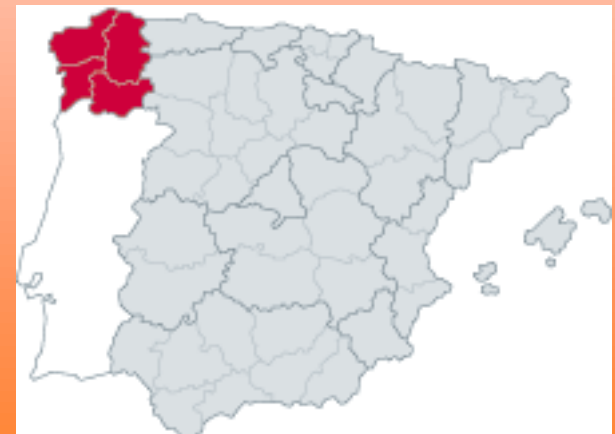
Relevant data

AEEG associates

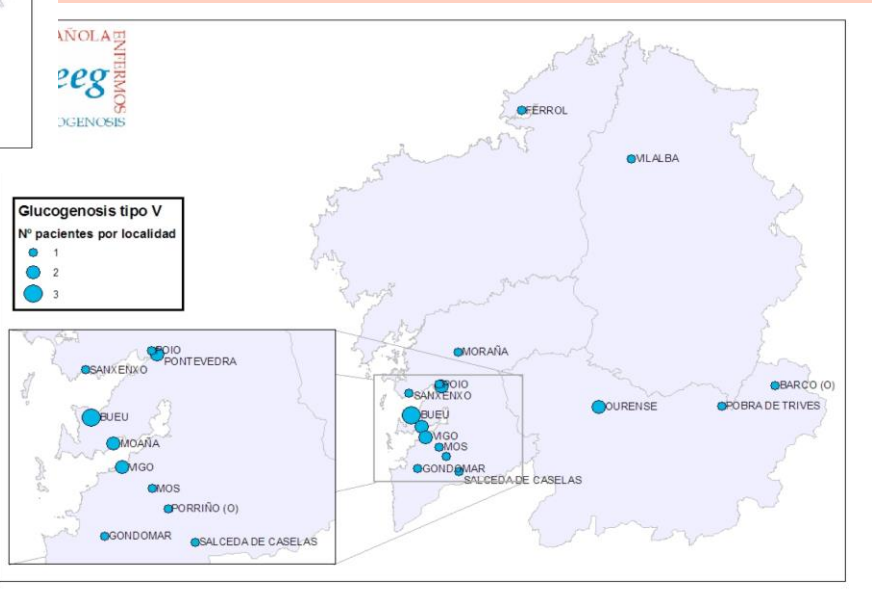
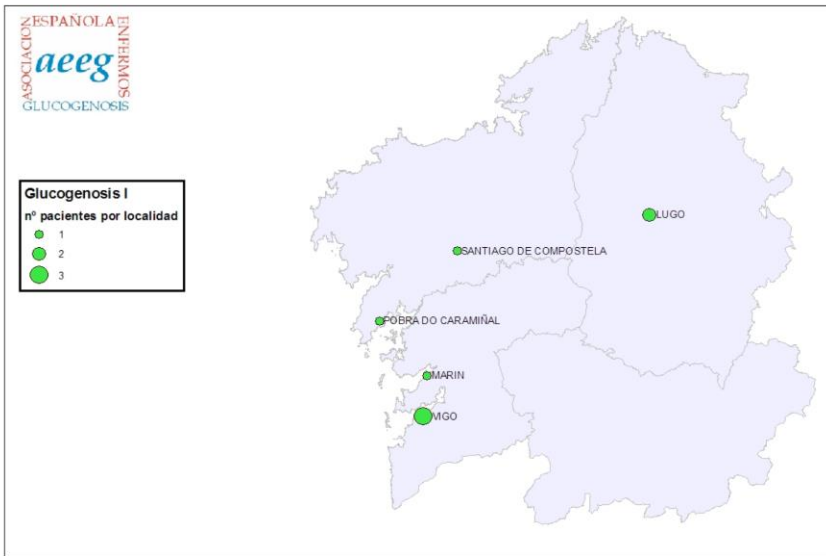


Projects

- 2007:
 - Glucogenosis en Galicia: Estudio de prevalencia y calidad de vida de pacientes afectados
(GSD in Northwestern of Spain: Study of prevalence and quality of life of affected patients)
 - Main researcher:
Molares Vila, Alberto



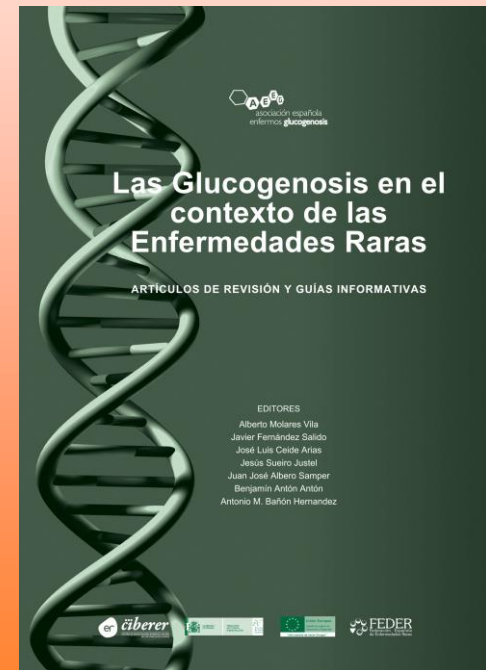
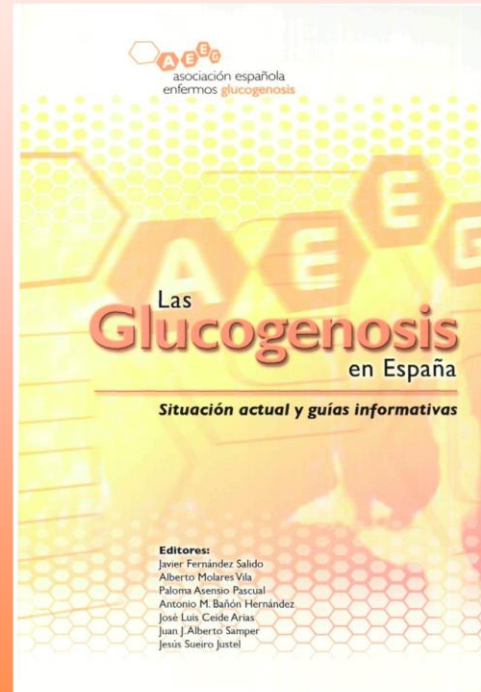
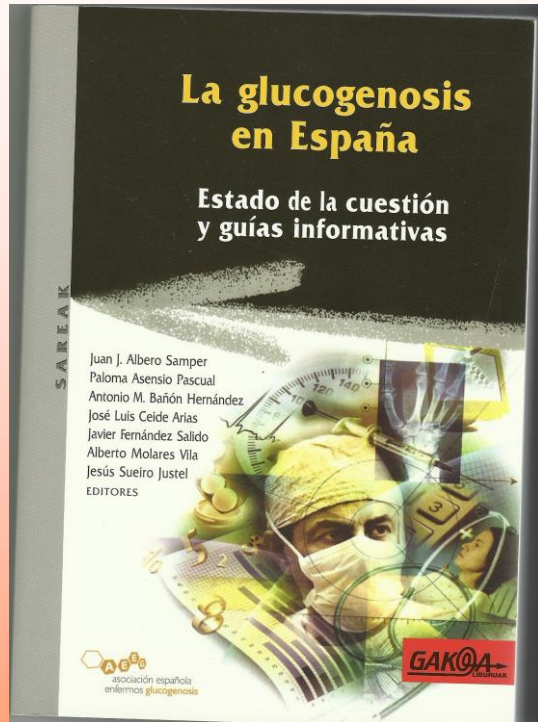
Projects



Assemblies/Congresses

- Only General Assemblies:
 - From 2000 to 2006 (Madrid)
- Congresses + General Assemblies:
 - 1st National: Seville (September 2007)
 - 2nd National: Madrid (June 2008)
 - 3rd National: Madrid (June 2009)
 - 1st International: Madrid (June 2010)
 - NEXT 2nd International: Burgos (June 2011)

Printed books



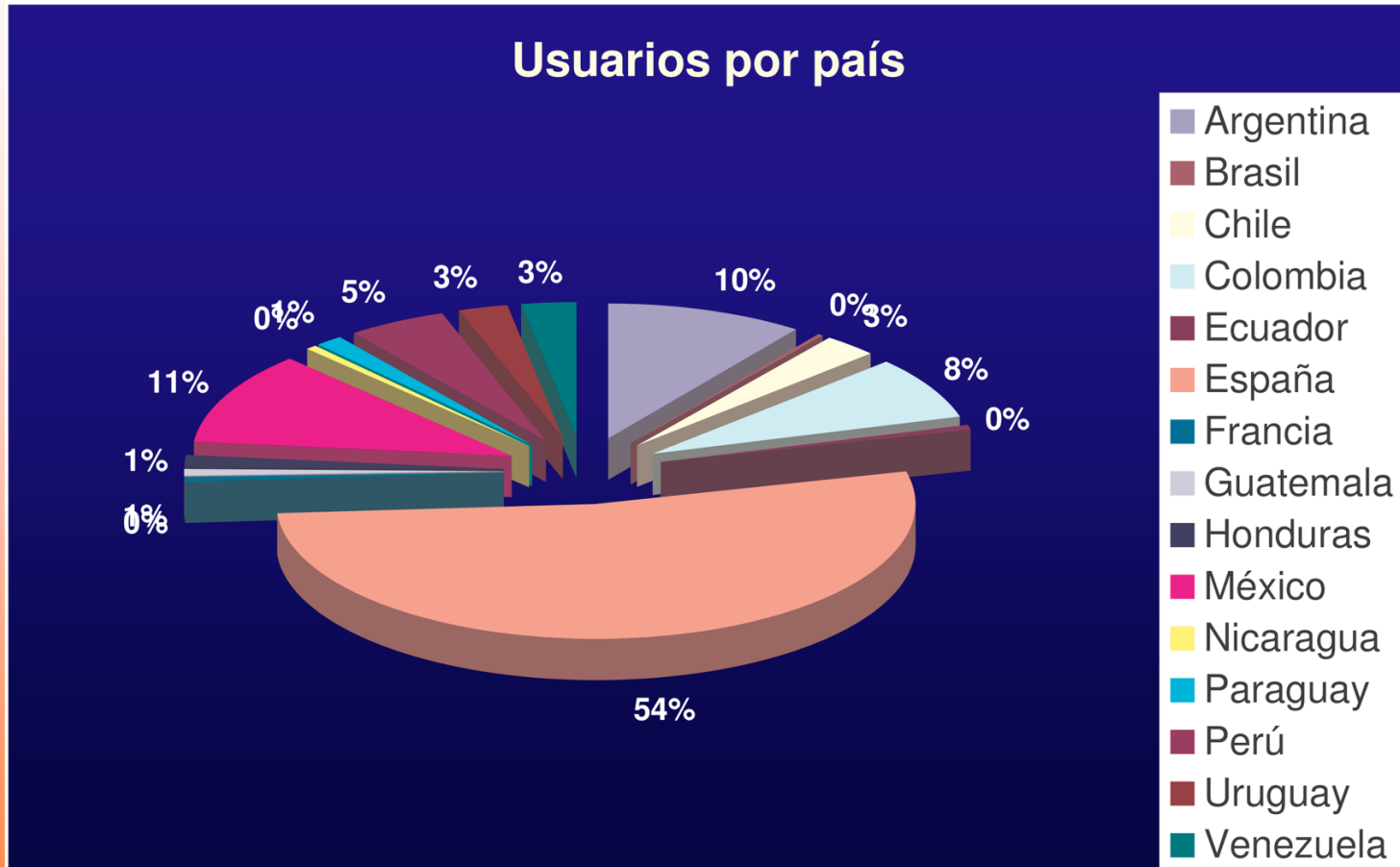
www.gluco-genosis.org

- Users: 10.011 (9/26/2010)
- Visits: 14.131 (9/26/2010)



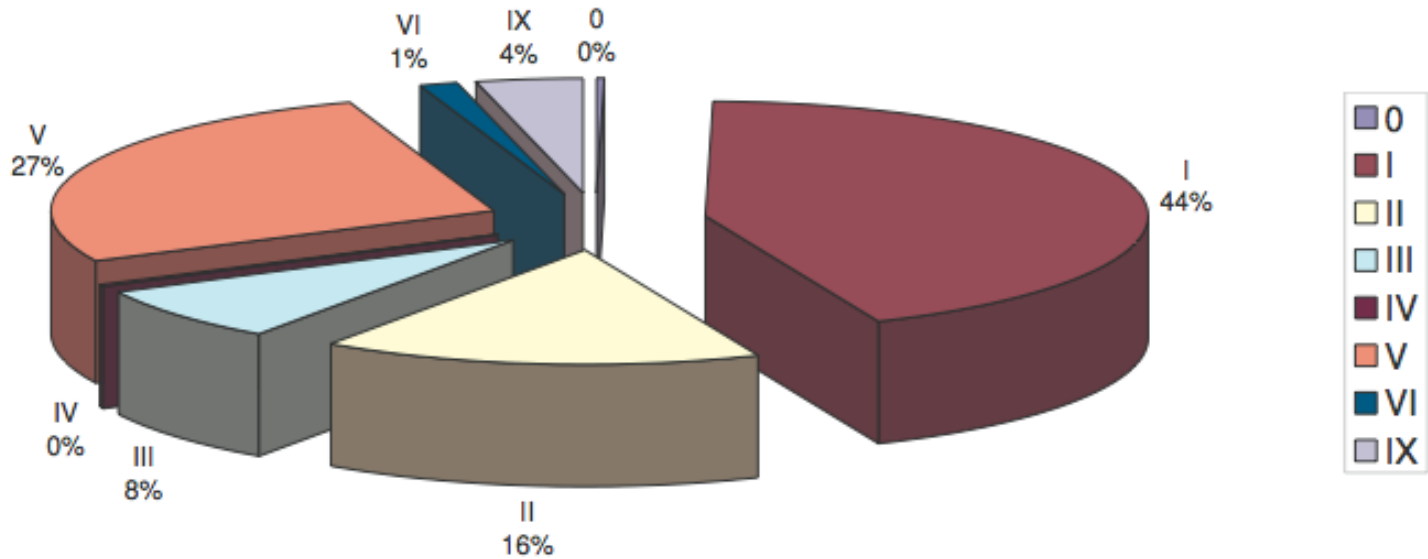
The screenshot shows the homepage of the AEEG website. At the top left is the logo, which consists of a hexagon followed by the letters 'A', 'E', 'E', and 'G' in orange boxes. Below the logo is the text 'asociación española enfermos gluco-genosis'. To the right of the logo is a login form with fields for 'Usuario' and 'Clave', a 'Recordar contraseña' checkbox, a 'Darse de alta' link, and an 'Entrar' button. A navigation menu on the left lists various sections: A.E.E.G, Glucogenosis, Estatutos, Junta Directiva, Guías publicadas, Foro, Noticias, Enlaces, Congresos-asambleas, Preguntas Frecuentes, Bibliografía, Traducciones, Alta socios, and Contacto. The main content area features a section titled 'Actualidad' with several news items, including 'Avances en terapia génica. Aplicaciones en Glucogenosis Tipo I', 'Noticias de salud: Pompe, una enfermedad difícil de diagnosticar', 'Registro de enfermedades raras del Instituto de Salud Carlos III', and 'El III Congreso de la AEEG en el boletín de la Universidad de Florida (Pag. 4). Artículo en inglés.' There is also a link to a 'Resumen Estudio Europeo sobre Glucogenosis tipo I'. On the right side, there is a section for a 'Convenio de colaboración entre la AEEG y el CIBERER' with the CIBERER logo, and a link to the 'II Congreso Internacional Glucogenosis' held in Burgos from June 3, 4, and 5, 2011.

www.glucogenosis.org



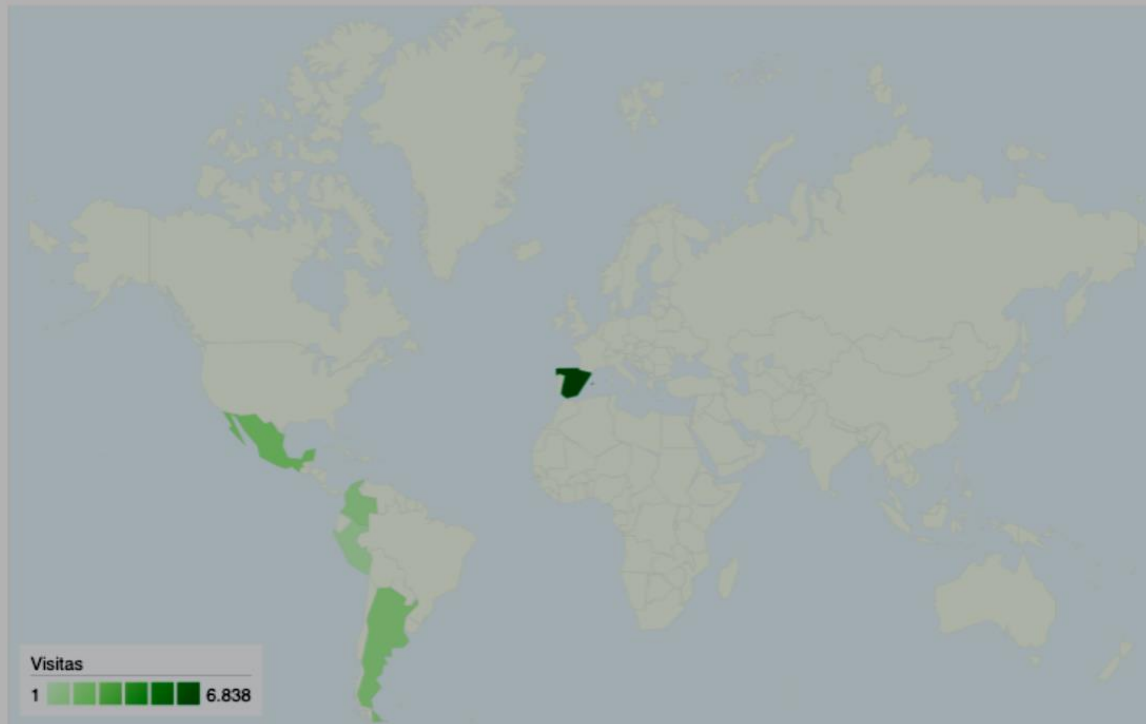
www.gluco-genosis.org

Distribución de tipos



www.gluco-genosis.org

Gráfico de visitas por ubicación



AEEG on Facebook

- Users: 599 (9/26/2010)
- Visits: 150 (9/26/2010)

The screenshot shows the Facebook interface for the AEEG profile. At the top, the Facebook logo is on the left, and the login section on the right includes fields for "Dirección de correo electrónico" and "Contraseña", with an "Entrar" button and a "No cerrar sesión" checkbox. Below the login section, a green "Regístrate" button is followed by the text "Asociación Española de Enfermos de Glucogenosis (AEEG) está en Facebook. Regístrate en Facebook para comunicarte con Asociación Española de Enfermos de Glucogenosis (AEEG)."

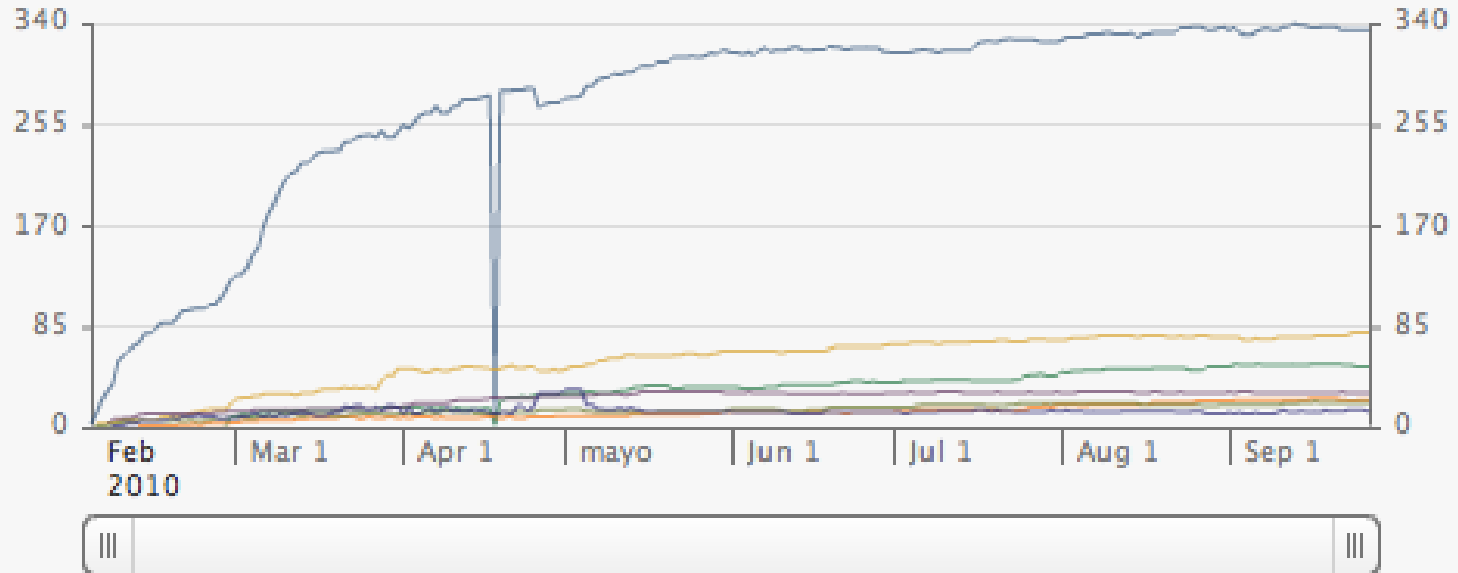
The profile header for "Asociación Española de Enfermos de Glucogenosis (AEEG)" features a profile picture of the organization's logo (a hexagon with 'AEEG' inside) and a "Me gusta" button. Navigation tabs include "Muro", "Información", "Fotos", "Foros", "Cuadros", "Eventos", and "Filtros".

A post by "Marúa José Gómez" is visible, with a profile picture of a woman. The post text reads: "Did you see the flash news about work-at-home-mom ? She makes \$89/hour working part time. Monthly 8795 \$ income from home... I just signed up and already earned 72 \$ this hour. It is really amazing... If you are looking for a work-at-home job, i suggest you to take a look at this press now... vtcrkitr". Below the text is a link titled "Today's Earnings: \$510.24" and another link titled "News Daily 7 - Work at home mom makes \$8,795/month working part-time from home" with the URL "www.daily-7-news-online.com". A short bio for Kelly Richards is also present: "Kelly Richards never thought that she would, until curiosity got the best of her and she filled out a simple online form. Before she knew it, she discovered her secret to beating the recession, and being able to provide for her family while at home with her three children."

At the bottom left, a box states "La Asociación Española de Enfermos de Glucogenosis, desde su constitución en 1999, ha querido ser, en la medida de sus posibilidades, un catalizador de las inquietudes e iniciativas de todos los colectivos afectados por estas enfermedades." Below this, a blue box indicates "A 599 personas les gusta esto".

AEEG on Facebook

- España Argentina México Uruguay Chile Venezuela
 Estados Unidos Colombia Perú Francia Reino Unido Costa Rica
 República Dominicana Guatemala Italia Alemania Ecuador
 Estonia Brasil Canadá



Current state of GSD research in Spain

National Research Network

Centre for Biomedical Network Research on Rare Diseases



“Una manera de hacer Europa”

National Research Network

<http://www.ciberer.es/index.php?lang=english>



The screenshot shows the homepage of the CIBERER website. At the top left is the CIBERER logo, which consists of a green circle with 'er' inside, followed by the word 'ciberer' in a stylized font. Below it is the text 'CENTRO DE INVESTIGACIÓN BIOMÉDICA EN RED DE ENFERMEDADES RARAS'. To the right of the logo are logos for the Spanish Government (GOBIERNO DE ESPAÑA), the Ministry of Science and Innovation (MINISTERIO DE CIENCIA E INNOVACIÓN), and the Instituto de Salud Carlos III. Further right are navigation links: Home | Intranet | Contact | Links | Search, and language options: Castellano - English.

Below the header is a horizontal menu with the following items: CIBERER (highlighted in green), SCIENTIFIC WORK, SCIENTIFIC AREAS, RESEARCH GROUPS, and RARE DISEASES.

The main content area features a large background image of a DNA double helix. Below this image is the text: 'Ayudas para el Fomento de la Dinamización del Entorno Tecnológico del Sistema Nacional de Salud. - 16º Premio Carmen y Severc'.

On the left side, there is a vertical sidebar menu with the following items: Programmes, Platforms, Calls, Calendar, Press Area, and Documents.

The central content area has a 'Welcome CIBERER' section. It includes a small photograph of a group of people at a conference. The text reads: 'Rare diseases, affecting around 3 million Spanish people, are a social and health problem of the first order. The Centre for Biomedical Network Research on Rare Diseases (CIBERER), a network structure set up at the initiative of the Instituto de Salud Carlos III, pools and furthers the excellence research done in this country with the aim of finding diagnoses and therapies for those affected as quickly as possible.'

On the right side, there is a 'CALENDAR' section with a list of events:

- 5º Encuentro Internacional de Biotecnología
29-Sep-10 - 01-Oct-10
- Master: Dirección y Gestión de la I+D+I en Ciencias de la Salud
30-Sep-10
- Introducción a la Investigación en Animales Modificados Genéticamente
30-Sep-10 - 01-Oct-10

At the bottom of the main content area, there is a 'NEWS' section with a green square icon on either side.

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- <http://www.ciberer-biobank.es/index.php?lang=en>

The screenshot shows the homepage of the CIBERER Biobank. At the top left is the 'cibererbiobank' logo with the tagline 'BANCO DE MUESTRAS BIOLÓGICAS DE ENFERMEDADES RARAS' and the 'cbk' logo. To the right is the 'ciberer' logo with the tagline 'CENTRO DE INVESTIGACIÓN BIOMÉDICA EN RED DE ENFERMEDADES RARAS'. Below these are logos for the 'GOBIERNO DE ESPAÑA', 'MINISTERIO DE CIENCIA E INNOVACIÓN', and 'Instituto de Salud Carlos III'. A navigation menu includes 'Documents', 'Catalogue', 'Links', 'Contact', and 'Samples Management'. A search bar and language flags (Spain and UK) are also present.

Home

- Home
- Structure
- Activities
- How it works
- Information Service
- FAQ's
- Cooperating institutions

CSISP
Centro Superior de Investigación en Salud Pública

BBMRI
Biobanking and

CIBERER BIOBANK



According to the definition given in the Biomedical Research Act 14/2007, a biobank is "a public or private non-profit-making establishment which has a collection of biological samples intended for diagnostic purposes or biomedical research and organised as a technical unit with criteria involving quality, order and allocation".

CIBERER BIOBANK, hereinafter **CBK**, is a public-non-profit-making biobank set up by the Centre for Biomedical Network Research on Rare Diseases (**CIBERER**) and located in La Fe Hospital in Valencia. Through being an integral part of the

CIBERER, the Biobank has the institutional backing of the Instituto de Salud Carlos III.

The **CBK** was founded with clear aspirations to excellence and differentiation. It has set out to

FAQ's

- Why send samples to the CBK?
- What is a Biobank?
- What is CIBERER?
- What are Rare Diseases?

News

- Se abre el plazo de admisión de 'Expresiones de interés' por parte de investigadores CIBERER para la donación de muestras

National Research Network

- Research groups on GSD



Group Card Investigation Lines Group Staff Publications

Dr. Eduardo Salido - U740

Type 2

Area: Enfermedades Metabólicas Hereditarias y Endocrinas (EMHE)

Departamento de Anatomía Patológica, Patología Molecular, Hospital Universitario de Canarias, Fundación Canaria Rafael Clavijo, Tenerife

Adress: Ofra s/n.La Cuesta

C.P.: 38320 La Laguna, Tenerife

Web:



Dr. Salido's group studies the development of molecular-based therapeutical strategies in primary hyperoxaluria. It uses first and foremost genetically modified mouse models to test cell, gene and enzyme therapy designs which could be used as concept evidence for new therapeutic approaches to this disease.

National Research Network

- Research groups on GSD



Group Card Investigation Lines Group Staff Publications

Dr. Eduard Gratacòs - U719

Type 2

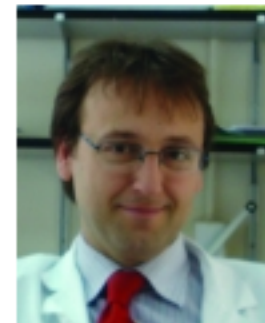
Area: Genética Clínica y Defectos Congénitos (GCDC)

Servicio de Obstetricia y Ginecología, Instituto de Investigaciones Biomédicas August Pi i Sunyer (IDIBAPS), Hospital Clínic y Provincial de Barcelona, Barcelona

Adress: C/ Sabino de Arana 1

C.P.: 08028 Barcelona, Barcelona

Web:<http://www.medicinafetalbarcelona.org>



A clinical and basic research team which undertakes several lines of research into early diagnosis, illness mechanisms and new therapeutic strategies of development disorders and congenital defects.

National Research Network

- Research groups on GSD



Group Card

Investigation Lines

Group Staff

Publications

Dr. Erwin Knecht - U721

Type 2

Area: Enfermedades Metabólicas Hereditarias y Endocrinas (EMHE)

Laboratorio de Biología Celular, Centro de Investigación Príncipe Felipe, Fundación

Centro de Investigación Príncipe Felipe (CIPF), Valencia

Adress: Av/ Autopista del Saler, 16-3 Camino de las Moreras

C.P.: 46012 Valencia, Valencia

Web:www.cipf.es



Dr. Knecht's group studies the mechanisms involved in the origin of disorders such as neuronal ceroid lipofuscinoses, Danon's disease and mitochondrial diseases, and cooperates with other CIBERER groups in studies on Lafora's disease and pigmentary retinosis

National Research Network

- Research groups on GSD



Group Card Investigation Lines Group Staff Publications

Dr. Antoni L. Andreu - U701

Area: Patología Mitocondrial (PM)

Unitat de Patologia Mitocondrial i Neuromuscular, Institut de Recerca Hospital
Universitari Vall d'Hebron, Institut Català de la Salut, Barcelona

Adress: Paseo Vall d'Hebron 119

C.P.: 08035 Barcelona, Barcelona

Web:<http://www.ir.vhebron.net>

Type 5



Study of the physiopathological mechanisms of mitochondrial diseases due to mutations in the mtDNA, using transmitochondrial cybrids. It also handles research into disorders due to mutations in the nDNA, particularly the type that affect nucleotide metabolism

National Research Network

- Research groups on GSD



Group Card Investigation Lines Group Staff Publications

Dr. Miguel Ángel Martín Casanueva - U723

Area: Patología Mitocondrial (PM)

Centro de Investigación, Hospital Universitario 12 de Octubre, Servicio Madrileño de Salud,
Madrid

Adress: Av/ de Córdoba s/n

C.P.: 28041 Madrid, Madrid

Web:

Type 5



Biochemical and molecular-genetic characterization of mitochondrial diseases and metabolic exercise intolerances. Study of (i) cellular pathophysiological consequences of mutations in mtDNA and nuclear genes related to the mitochondrial respiratory complexes; (ii) role of the assembly of the respiratory complexes in mitochondrial pathology; (iii) genotype modulators for type V glycogen storage disease phenotype; and (iv) the neurodegenerative process in ELA by using cellular and animal models.

National Research Network

- Research groups on GSD



Ficha **Líneas de Investigación** Personal Grupo Publicaciones

Dr. Francesc Cardellach López - U722

Area: Patología Mitocondrial (PM)

IDIBAPS(Patología Mitocondrial), Instituto de Investigaciones Biomédicas August Pi i Sunyer (IDIBAPS), Hospital Clínic de Barcelona, Barcelona

Dirección: Laboratorio 413.Medicina Interna.C/ Villarroel 170

C.P.: 08036 Barcelona, Barcelona

Web: <http://www.idibaps.ub.edu/cat/home.php>

Type 5



Patología mitocondrial, tanto primaria como secundaria (enfermedades neurodegenerativas, intoxicaciones, efectos secundarios de terapias, base mitocondrial de procesos fisiológicos o de enfermedades comunes como la sepsis). Aparte del estudio mitocondrial, este grupo investiga otras miopatías (inflamatorias, metabólicas o distrofias) en adultos.

National Research Network

- Research groups on GSD



Unidad Enfermedades Neuromusculares

Servei de Neurologia
Hospital de la Santa Creu i Sant Pau
C/ Pare Claret, 167
08025 Barcelona
Spain

Tel. +34 935 565 977
Fax: +34 935 565 602
E-mail: 8082ifa@comb.cat

Investigadora Principal

Isabel Illa Sendra

Type 5

International Meeting GSD Associations
Milan, October 2nd-3rd, 2010

National Research Network



Research groups on hepatic GSDs (type I, III, VI and IX)???

National Research Network

- Research groups on GSD



Group Card Investigation Lines Group Staff Publications


Hepatics

Dra. Antonia Ribes - U737

Area: Enfermedades Metabólicas Hereditarias y Endocrinas (EMHE)

Servicio de Bioquímica y Biología Molecular , Institut de Bioquímica Clínica, Servicio de Bioquímica y Genética Molecular, Hospital Clínic y Provincial de Barcelona, Barcelona
Adress: Edificio Helios III planta Baja C/ Mejía Lequerica s/n
C.P.: 08028 Barcelona, Barcelona

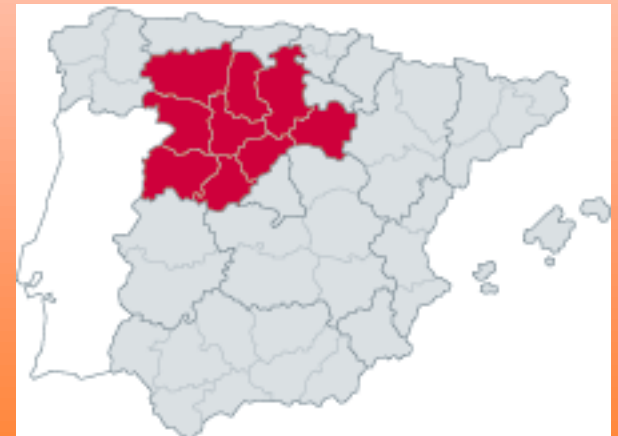
Web:



The main line of research and diagnosis tackled by this group is "Inherited Metabolic Diseases" (IMD). We have carried out translational research and applied new technologies in order to improve diagnosis and prevention of these diseases. Consequently we currently cover a wide diagnostic range: disorders of the metabolism of amino acids, organic acids, pyruvate, fatty acids, galactose, glycosylation, cholesterol, as well as peroxisomal, mitochondrial and lysosomal diseases. Our laboratory is a reference centre for studying IMD; it has implemented the ISO 9001-2008 quality system and participates in external quality programmes ERNDIM, CDC and AECNE. As part of the general line of IMD, our recent specific lines of research are based on "in vitro" therapeutic approaches: CoQ10 Deficiency, Cerebral Creatine Deficiency, mtDNA Depletion, ERAB or MHBD Deficiency, Type C Niemann-Pick Disease and X-linked Adrenoleukodystrophy.

National Research Network

National Reference Care Centre for People with Rare Diseases and their Families (Burgos)



National Research Network

National Reference Care Centre for People with Rare Diseases
and their Families

http://www.imserso.es/creer_06/index.htm



The screenshot shows the website for the National Reference Care Centre for People with Rare Diseases and their Families (Creer). The header includes the logos of the Spanish Government (GOBIERNO DE ESPAÑA), the Ministry of Health and Social Policy (MINISTERIO DE SANIDAD Y POLÍTICA SOCIAL), and IMSERSO. The main navigation menu includes Home, Contact Us, The RDs, Orphan drugs, Frequently questions, News, and Agenda. The main content area features a large banner with a photograph of the building and the Creer logo. Below the banner is a description of the center's role. To the right, there are several informational boxes: 'Premios Imerso Infanta Cristina 10', 'Electronic Headquarters', '901 10 98 99 INFORMACIÓN DEL IMSERSO', and a quote 'Combatir el calor está en tus manos'. At the bottom right, there is a calendar for September 2010.

Castellano Català Euskara Galego Valencià **English** Français

GOBIERNO DE ESPAÑA
MINISTERIO DE SANIDAD Y POLÍTICA SOCIAL
IMSERSO
creer
CRE Enfermedades Raras (Creer)
Burgos

Home | Contact Us | The RDs | Orphan drugs | Frequently questions | News | Agenda

Information Sections

- › The Centre
- › Service Users
- › Direct Care Services
- › Reference Services
- › Resources
- › Documentation
- › Administrative Procedures
- › The professional's blog
- › Creer's Chat
- › Links

Direct Access

Latest News

Premios Imerso Infanta Cristina 10

Electronic Headquarters

901 10 98 99 INFORMACIÓN DEL IMSERSO

Combatir el calor está en tus manos

Septiembre 2010

Mo	Tu	We	Th	Fr	Sa	Su
		1	2	3	4	5
6	7	8	9	10	11	12
13	14	15	16	17	18	19

National Research Network

National Registry of Rare Diseases

<https://registorraras.isciii.es/comun/inicio0.aspx>

GOBIERNO DE ESPAÑA **MINISTERIO DE CIENCIA E INNOVACIÓN** **Instituto de Salud Carlos III** **Registro de Enfermedades Raras**

Solicitud de Inclusión

::Login

Usuario:

Contraseña:

Entrar

Logout

Usuario anónimo
Para acceder debe autenticarse primero.

[Manual de Usuario](#)

[Preguntas Frecuentes](#)

Presentación

Para registrarse pulse siguiente botón. **Registro Web**

Para registrarse por correo postal pulse siguiente botón. **Registro Correo Postal**

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 (Consorcio de Investigación Biomédica en Red de Enfermedades Raras).

El objetivo de este registro es proporcionar a los profesionales de los sistemas de salud, investigadores y colectivos de pacientes y familiares afectados de una enfermedad rara, un mayor nivel de conocimiento acerca del número y distribución geográfica de los pacientes afectados por éstas enfermedades, con el objetivo de fomentar la investigación sobre las mismas, aumentar su visibilidad y favorecer la toma de decisiones para una adecuada planificación sanitaria y una correcta distribución de recursos.

El registro se sustenta desde el punto de vista legal en la orden publicada en el B.O.E de junio de 2005, (ORDEN SCO/1730/2005, DE 31 DE MAYO), donde se establecieron los criterios de creación y

Aprobación del Comunicado de la Comisión Europea sobre Enfermedades Raras
17/11/2008-31/12/2011
[Enlace Externo](#)

RECOMENDACIÓN DEL CONSEJO relativa a una acción en el ámbito de las enfermedades raras
11/06/2009-31/12/2011
[Enlace Externo](#)

Estrategia Nacional en Enfermedades Raras
30/10/2009-30/06/2011
[Enlace Externo](#)

Acuerdo entre EURORDIS y NORD
30/10/2009-01/10/2010
[Enlace Externo](#)

National Research Network

National Registry of Rare Diseases

Pacientes registrados por Correspondencia OMIN

Mostrando 53 registros

<u>Id Oiml</u>	<u>Descripción</u>	<u>Numero de Pacientes</u>
261600	Fenilcetonuria (PAH): Fenilalanina hidroxilasa, Deficiencia de: Fenilalanina monooxigenasa, Deficiencia de	1
309550	Fragile site mental retardation 1 (FMR1)	35
309548	Fragile site, folic acid type, rare, Fra(X)(q28) (FRAXE)	35
229300	Friedreich ataxia 1 (FRDA1)	26
230500	Gangliosidosis GM1	1
232200	Glucogenosis Tipo Ia: Actividad hidrolítica de la glucosa-6-fosfatasa, Deficiencia de la	8
232240	Glucogenosis Tipo Ib	8
232300	Glucogenosis Tipo II: a-glucosidasa ácida, Deficiencia de	11
232400	Glucogenosis Tipo III: Amilo 1,6-glucosidasa, Deficiencia de	1
232600	Glucogenosis Tipo V: Glucógeno fosforilasa, Deficiencia de	6
308000	Hipoxantina guanina fosforribosiltransferasa, Deficiencia de	2
143100	Huntington chorea (HD)	3
146000	Hypochondroplasia (HCH)	3
245200	Krabbe, Enfermedad de	2
250100	Leucodistrofia metacromática (MLD)	5

[1](#) [2](#) [3](#) [4](#)



asociación española
enfermos **glucogenosis**

Thank you very much!

Asociación Española de Enfermos de
Glucogenosis (AEEG)

www.glucogenosis.org

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