

/ OPINA

Los pacientes opinan en Guía metabólica, accede a nuestra encuesta y ayúdanos a mejorar. ¡Damos tu opinión e ideas!

/ NUESTRAS RECETAS

En La Cuineta tenemos sobrosas recetas adaptadas para la alimentación restringida en proteínas, grasas o glúcidos, y también puedes enviarnos tus propias recetas para compartirlas.

/ CONSEJOS

A parte de las experiencias de padres y profesionales incluímos recomendaciones útiles que pueden ayudar en el día a día de las familias, en la escuela, en los deportes o en la alimentación.

INICIO DE SESIÓN

Usuario:
 Contraseña:

 ¿No tienes cuenta? Solicita una nueva contraseña

/ Comentarios recientes

- HSD Dra. M Serrano en Galactosemia
- HSD Dra. M Serrano en Glucocorticoides
- mina en Galactosemia
- HSD Natalia Ego en Huesos duros: valores de calcio de suero y Quercitas
- enrique sequeiros en Glucocorticoides

/ Información médica



¿Qué sabes del boniato?
 16/11/2011
 El origen del boniato se sitúa en América central y Sudamérica, donde se los conoce por varios nombres, como camote o batata. Hasta el siglo XVI no llegó a Europa y de allí se extendió por todo el mundo debido a su valor nutritivo y su buen gusto.

Actualidad médica alimento del mes boniato [Actualidad](#)



Save the PKU Planet!
 16/11/2011
 El pasado 11 de noviembre se celebró en el Complejo Administrativo Triunfo de la Universidad de Granada una Jornada Internacional médico-sanitaria sobre la fenilcetonuria (PKU) dentro de la cual se presentó el videojuego "Save the PKU Planet".

Actualidad médica fenilalanina fenilcetonuria juego PKU [Actualidad](#)

/ Quién está en línea

Actualmente hay 0 usuarios y 4 invitados en línea.

/ Glosario

globos pálidos
 Una de las estructuras de los núcleos de la base que tiene importantes funciones en el control del movimiento. ...



/ Eventos



XIV Reunión para pacientes y familiares con PKU y OTM del Hospital Sant Joan de Déu
 26/11/2011
 La Unidad de seguimiento PKU y OTM del Hospital Sant Joan de Déu de Barcelona organiza el 26 de noviembre la XIV Reunión para pacientes y familiares con PKU y Otros Grupos del Metabolismo, que se celebrará en el Auditorio del Edificio Docente del centro.

Eventos OTM PKU [Actualidad](#)

/ Siguenos en



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/ Buscador de recetas

Dieta controlada en:

Proteínas Glúcidos Grasas

Platos:

1º plato 2º plato Postre

Aperitivo Salsas Panes y Massas Fiesta

Ingrediente:

/ Recetas

Glúcidos **Proteínas** **Grasas**

Salsa bechamel con bebida de soja
 HSD:BOI

Láminas de boniato con salteado de setas y refrito con carne
 Usuario: Lali

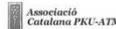
Croquetas de pollo
 Usuario: Lali

[Consumir BROSKE](#)

/ Envía tu receta

¿Tienes alguna receta deliciosa que quieres compartir?
 Inicia sesión o regístrate, envíanosla y nuestro equipo médico valorará su publicación en La Cuineta.

Un proyecto de:



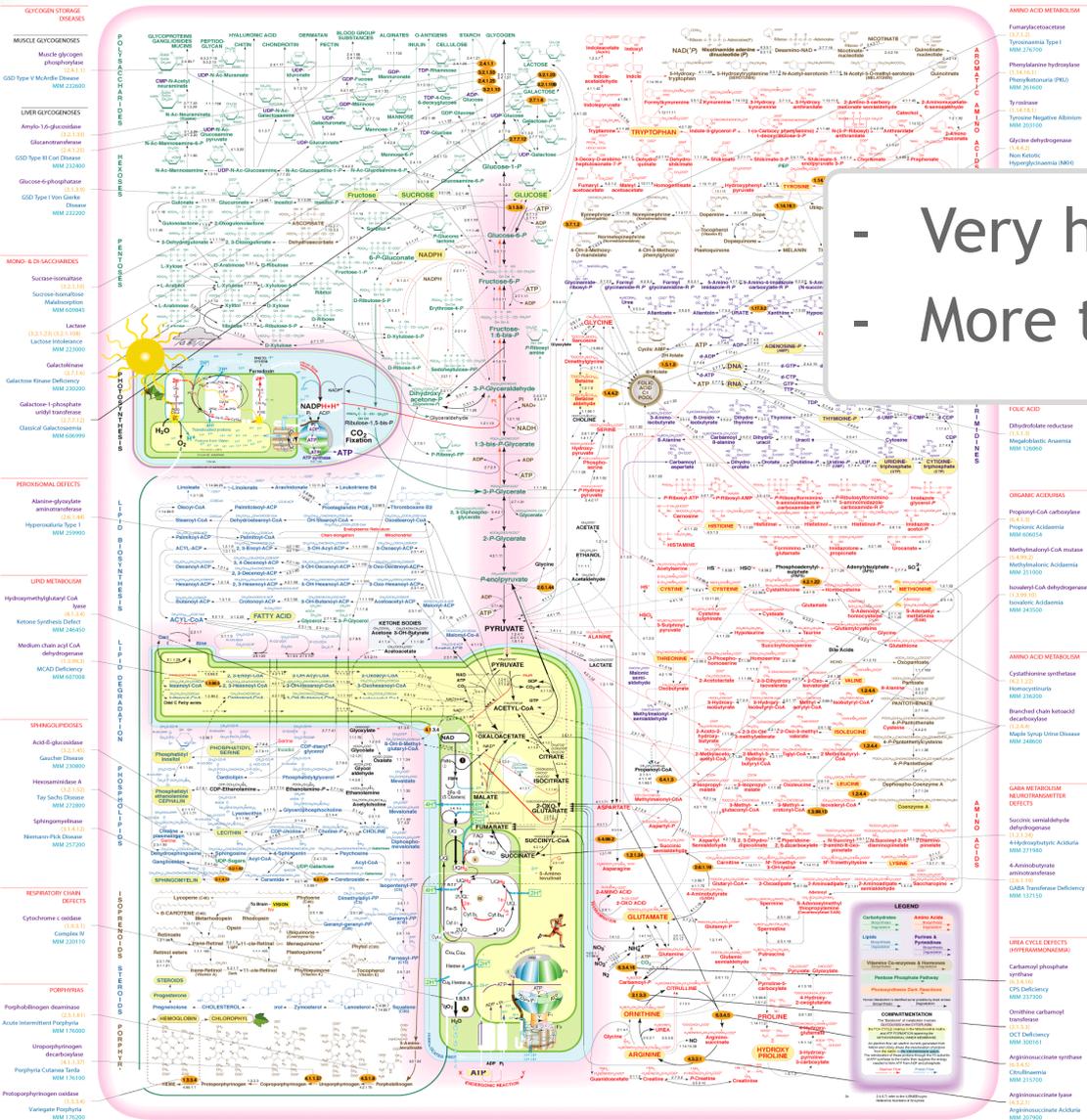
Con el apoyo de:



m3tabólica guía

Merche Serrano, MD PhD (@serranogimare)
 Centre for Biomedical Network Research on Rare Diseases (CIBERER), ISCIII and Unidad de Enfermedades Metabólicas.
 Hospital Sant Joan de Déu.
 Barcelona, Spain,

Inborn Errors of Metabolism



- Very heterogeneous group
- More than 500 diseases

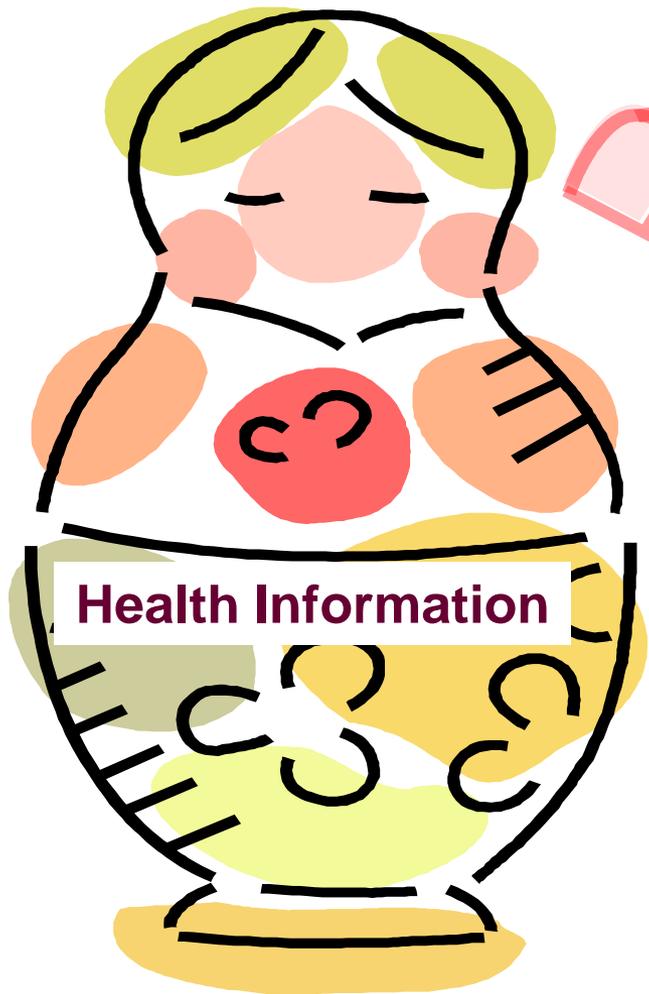


Addition

1. Lack of information



1. Resources



Information for
IEM



Easy-to-
understand
information?

AIM

To facilitate the access to
information

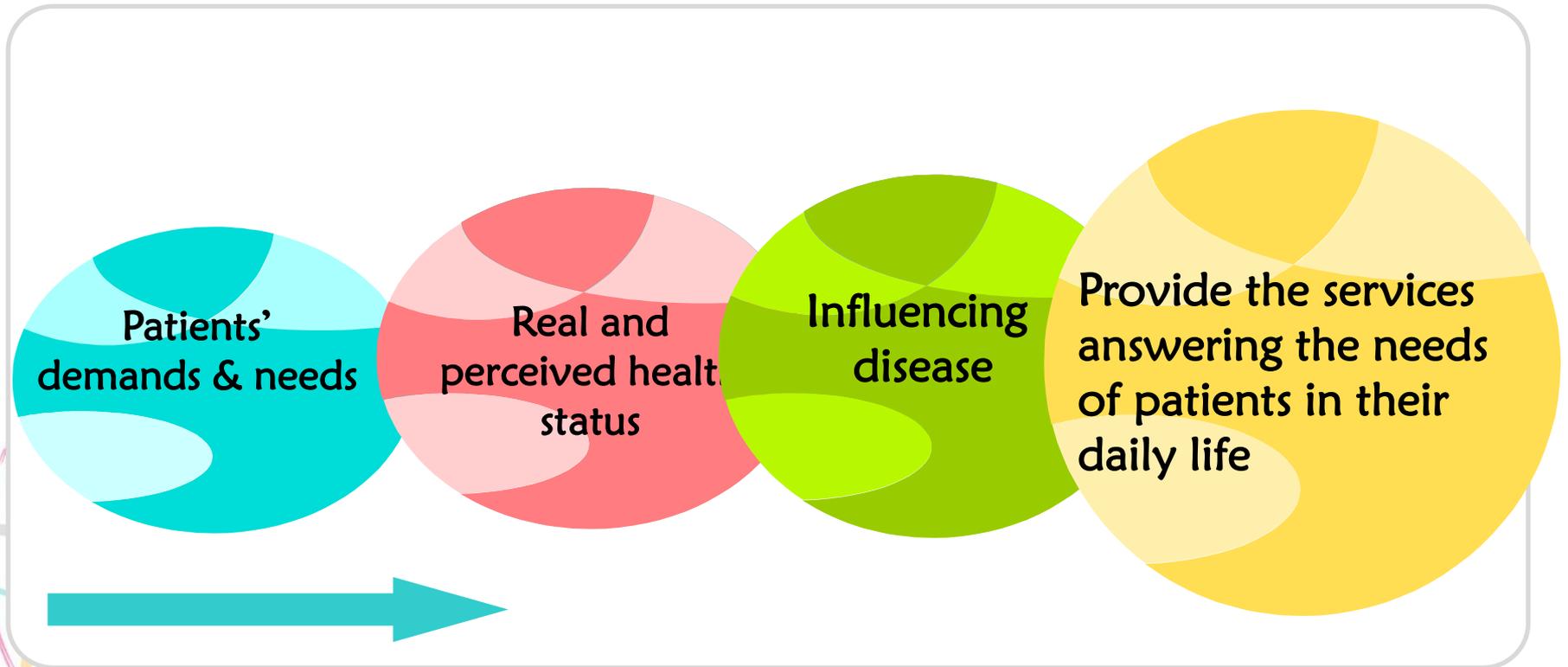
- Quantity
- Quality (*easy jargon*)

Addition

1. Lack of information
2. Classic model of medicine is not enough.



2. Patients' Demands & Needs



Addition

1. Lack of information
2. Classic model of medicine is not enough.
3. The effectiveness of Web-based interventions on patient empowerment in chronic diseases has recently been proven.
4. Online support groups reduce feelings of loneliness and social isolation



“Social Media is almost the only way to socialize in RD, particularly in ultraRD” [@armayones](#)

Addition

AIM

To create an interface for people involved in IEM

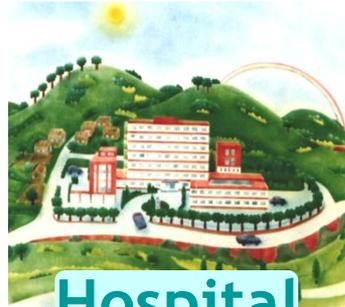
- contact with professionals and other similar patients,
- offering a platform for developing support groups.



“Social Media is almost the only way to socialize in RD, particularly in ultraRD” [@armayones](#)

Addition

1. Lack of information
2. Classic model of medicine is not enough.
3. The effectiveness of Web-based interventions on patient empowerment in chronic diseases has recently been proven.
4. Online support groups reduce feelings of loneliness and social isolation



guía
m3tabólica

Methods

We developed a frequently-updated 2.0 website in Spanish for IEM patients

Regístrate | Accesibilidad | Mapa web

Buscar:

guía
metabólica

Sant Joan de Déu
HOSPITAL MATERNOINFANTIL - UNIVERSITAT DE BARCELONA

ECM CONSEJOS LA CUINETA APRENDER JUGANDO INFORMACIÓN ¿QUIENES SOMOS?

OPINA

Opina

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NUESTRAS RECETAS

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GEO-RECURSOS

¿Sabes dónde comprar alimentos especiales, farmacias que dispongan de medicamentos para **ECM** o restaurantes y hoteles sensibles a vuestras necesidades? Recomienda tus propios Geo-Recursos.

INICIO DE SESIÓN

Usuario:

Contraseña:

INICIAR SESIÓN

[Crear nueva cuenta](#)
[Solicitar una nueva contraseña](#)

Comentarios recientes

MARI en [Homocistinuria con aciduria metilmalónica \(CblC\)](#)

HSJD Natàlia Egea en [Homocistinuria clásica](#)

HSJD Mei García en [Defectos de la \$\beta\$ -oxidación](#)

dicampos en [Defectos de la \$\beta\$ -oxidación](#)

HSJD Dra. M Serrano en [Homocistinuria con aciduria metilmalónica \(CblC\)](#)

Información médica

CDG

El I Encuentro Luso-Ibérico de los Defectos Congénitos de la Glicosilación homenajea al Profesor Jaak Jaeken

03/11/2011

Las familias con pacientes afectos de CDG de 3 países (España, Portugal y Francia), así como a diversos facultativos (clínicos, bioquímicos y genetistas) involucrados en los CDG, se reunieron en Barcelona el pasado noviembre, presididos por el **Profesor Jaak Jaeken (Departamento de Pediatría, Universidad de Leuven, Bélgica)**, quien describió estas enfermedades hace 30 años.

[Actualidad médica](#) [CDG](#) [defecto congénito de la glicosilación](#) [síndrome CDG](#)

Siguenos en

[f](#) [RSS](#) [YouTube](#)

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GUARDAR

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Quién está en línea

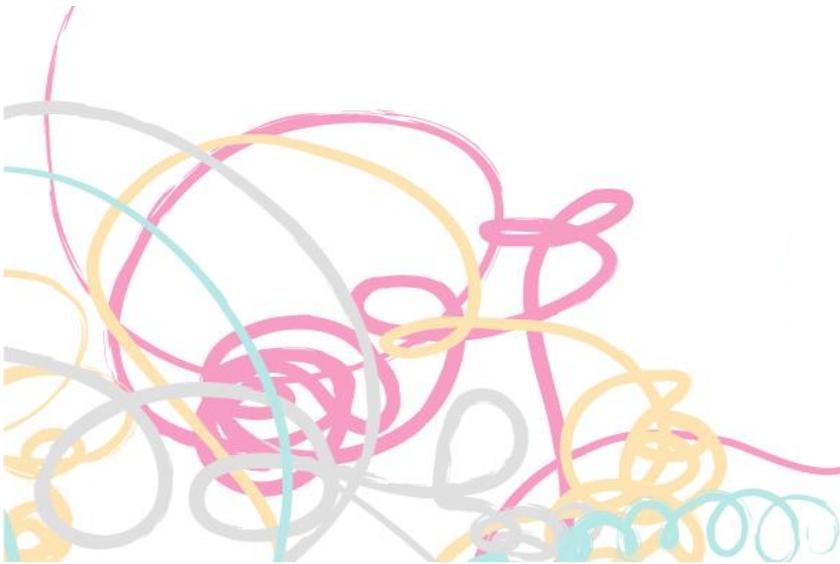
Manejo de la disfagia en casa

26/10/2011

Algunas enfermedades metabólicas vienen acompañadas de **difagia** que normalmente

Target audience

Patients, patients' families,
patients' associations, teachers,
friends, physicians, nurses, researchers...



Target audience

Considering that Spanish is used on the website and based on the incidence of IEM.

Target audience



62.000 people

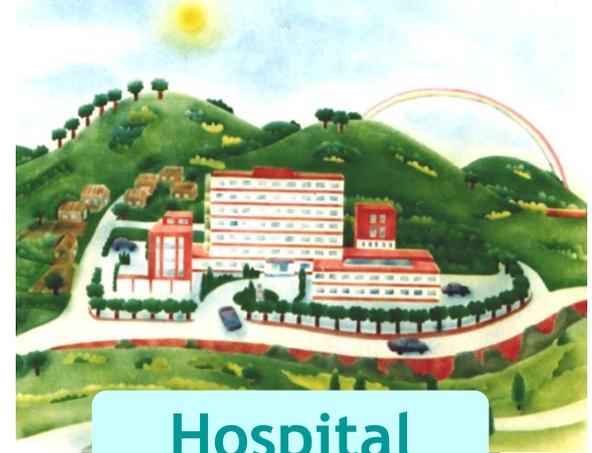


Mediators

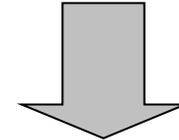
E-PATIENTS



E-CAREGIVERS



Hospital



Medical Staff



Mediators

E-CAREGIVERS

83%



1. Healthy people, 30-to-50 years old

2. Strong affection

3. Disconcerting situation of a child suffering a chronic illness



More participative, involved and committed



Questions

- ❖ **How platform allowed for interaction between researchers, patients, and families on medical and nutritional questions?**
- ❖ What structures were put in place?
- ❖ What resources or time was needed from your staff?
- ❖ What was the benefit to families or patients?
- ❖ What are the results so far?
- ❖ What have you learned along the way?
- ❖ What are your plans for the future?
- ❖ Using social media to stimulate community building between specialists and patients, what tools are needed?

[Inicio](#) » [Enfermedades](#) » [Homocistinuria con aciduria metilmalónica \(CbIC\)](#)



Homocistinuria con aciduria metilmalónica (CbIC)

Es un error congénito del metabolismo de la vitamina B12 o cobalamina, que causa la acumulación en plasma, orina y tejidos de homocisteína y ácido metilmalónico.

Clasificación: Metabolismo intermedio » Metabolismo de aminoácidos
Gen: MMACHC

Dieta controlada en **Proteínas**

ÚNETE A

Somos 130 usuarios

PARTICIPA

Hay 94 participaciones

La enfermedad

- ¿Qué es una homocistinuria con aciduria metilmalónica?
- ¿De dónde proceden la homocisteína y el ácido metilmalónico?
- ¿Por qué se produce una homocistinuria con aciduria metilmalónica?
- ¿Qué ocurre en el caso de un niño/a que nace con una homocistinuria con aciduria metilmalónica?
- ¿Cómo se diagnostica una aciduria metilmalónica con homocistinuria?
- ¿Qué hay que hacer para evitar las consecuencias de una homocistinuria con aciduria metilmalónica?

Consejos

Sugerencias, ideas y recomendaciones de los expertos.

La cuineta

Información, recursos y recetas de nutrición.

- Recetas
- Envía tu receta
- La importancia de la alimentación
- Recursos de nutrición

Aprender jugando

El espacio para los peques con cuentos y juegos.

Homocistinuria con aciduria metilmalónica (CbIC)

/ Comentarios

L CISTINA

Enviado por majonieto26 el Lun, 14/05/2012 - 16:12.

DRA. SERRANO
AGRADEZCO INFINITAMENTE SUS RESPUESTAS. MI HIJO VITTO GRACIAS A DIOS SE ENCUENTRA MUCHO MEJOR. ESTUVO MUY DELICADO. AL DIA DE HOY, EN CONSTANTE MEJORA.
PODRIA USTED INFORMARME SOBRE EL USO DE LA L CISTINA PARA BAJAR EL NIVEL DE HOMOCISTEINA? ES MUCOLITICO? LE AGRADEZCO LA INFORMACION! SALUDOS!

[responder](#)

Cist(e)ina

Enviado por HSJD Dra. M Serrano el Jue, 17/05/2012 - 23:02.

Hola,
Hay algunos estudios que evalúan la relación de la cistina en plasma y los valores de homocisteína y han visto que, en casos severos de homocistinuria, la administración de cistina puede resultar beneficiosa. Sobre todo veían esta relación cuando la cistina en plasma se encontraba por debajo de 170 micromol/L, ahí era cuando se notaba un incremento marcado de los valores de homocisteína. Estos estudios se han realizado en pacientes con defectos de Cbl-C, MTHFR...es decir homocistinurias no clásicas.
Adjunto información sobre el artículo más reciente por si os puede resultar de utilidad. Pero, como siempre, debe ser un médico el que asesore sobre la necesidad de iniciar la medicación, así como la dosificación y la pauta de administración de la misma.
<http://www.ncbi.nlm.nih.gov/pubmed/17186416>

Recibe un cordial saludo.

Dra. M Serrano
Neuropediatra. CIBERER. Unidad de Enfermedades Metabólicas

Hospital Sant Joan de Déu

Adjuntos

Tríptico de la homocistinuria con aciduria metilmalónica (CbIC) [400.82 KB]

/ HSJD Dra. M Serrano

- [Mi cuenta](#)
- [Terminar sesión](#)

/ Síguenos en



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Usuario:
HSJD Dra. M Serrano

[DARSE DE BAJA](#)

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Envía tus recetas

Comment field

Medical/nutritional
questions

Share experiences
among families

Supervised by our
Medical/Nutritional
staff



Comment Field: Analysis of users' questions



We had, on average, 12 weekly consultations that have been handled by the nutritional/medical staff, involving all 970 comments

50%

- 22.1% Information about their IEM (diagnostic tools, treatment...)
- 10.2% Doubts about other transitory or concomitant situations and their treatment
- 16.6% Doubts about dietary treatments

50%

- 17.5% Requests to contact other patients/caregivers
- 14.8% Messages exclusively to thank and express feelings
- 13.0% Patients' experiences and reflections, tips or recommendations
- 5.8% Questions about practical issues of the daily life (travel, shops...)

Comment field



Equilibrium between:

- those comments regarding educational issues and
- those related to Guiametabolica.org function as a support group.

The first type of comments (**educational issues**) would probably increase users' knowledge about the management of the disease, an essential element for a normal process of patient empowerment.

The second type of comments (**Guiametabolica.org as a support group**), enhance their emotional resources and stability, which is indispensable too.

Questions

- ❖ How platform allowed for interaction between researchers, patients, and families on medical and nutritional questions?
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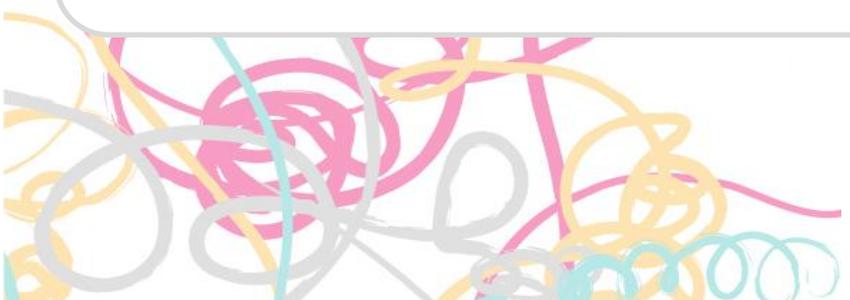
Our team

Guiametabolica.org team:

- (1) one nutritionist,
- (2) one specialist in clinical **biochemistry**,
- (3) one child **neurologist**,
- (4) one **mother** representing families and Patients' Associations,
- (5) one **journalist** specialized in social media and the Internet,
- (6) one expert in online audio-visual media and webmaster.



Time needed?



Content

- (1) Scientific information in simple language about 58 IEMs.
- (2) More than 90 tips and recommendations for daily life
- (3) Translated abstracts of more than 400 articles with scientific contents and medical advances, as well as monographic articles about medical issues.



Content

- (4) 195 specific recipes for controlled diets (proteins, carbohydrates, fat)
- (5) 16 complete menus

Coca de San Juan



-  Dieta controlada en **Proteínas**
-  Fiesta
-  Fundación ALICIA

Proteínas Fiesta azúcar fruta confitada harina Harifen huevos de codorniz
leche PKU Lecite levadura mantequilla [Receta](#)

Turrón de cereales bajos en proteínas y chocolate



-  Dieta controlada en **Proteínas**
-  Contenido en proteínas **Bajo**
- Kcal** Hipercalóricas
-  Postre
-  HSJDBCN

Proteínas Bajo Postre Fiesta canela cereales bajos en proteínas chocolate
frutas confitadas harina baja en proteínas [Receta](#)

Briwat salada



-  Dieta controlada en **Proteínas**
-  Aperitivo
-  Usuario: Familia Daoudi El Morabit

Proteínas Aperitivo Fiesta aceitunas cebolla fideos chinos harina baja en
proteínas papel de arroz pimientos setas [Receta](#)

Briwat dulce con arroz

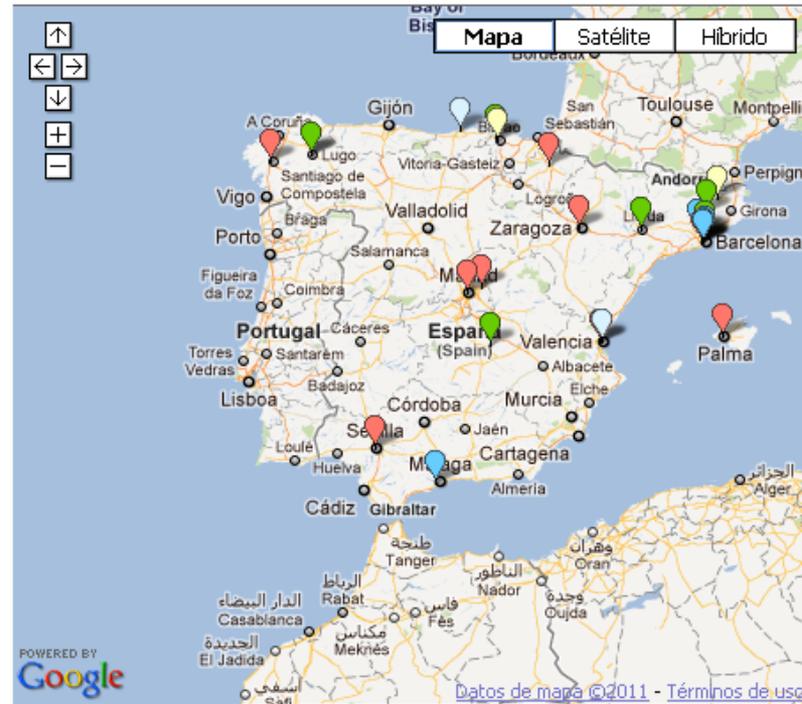


-  Dieta controlada en **Proteínas**
-  Contenido en proteínas **Bajo**
-  Postre
-  Usuario: Familia Daoudi El Morabit



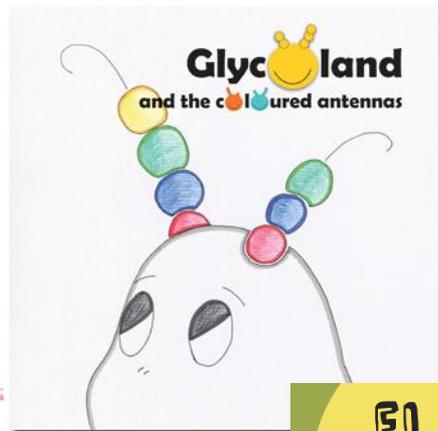
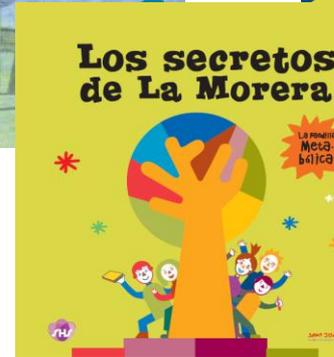
Content

Geolocated resources



Content

Stories



Content

Games

<http://www.fenilcetonuria.es/cocinaPKU.htm>



Newsletter & Social Networks

Every month, a newsletter is published and sent to the registered user database (n>800)

Guiametabolica.org has three different spaces in Social Networks like Facebook, Twitter and Youtube.

Octubre 2011



¿Sabías que...?

Hay casi 50 Errores Congénitos del Metabolismo catalogados como Defectos de la Glicosilación de Proteínas (CGD). Todos ellos han sido descritos en las 2 últimas décadas, lo que quiere decir que ha habido un gran avance en el diagnóstico de los ECM, pero que realmente queda mucho por descubrir. No te pierdas la [entrevista al Profesor Dr. Jaak Jaeken](#), el primer científico en describirlos.

Información



Guía metabólica: los pacientes opinan

29/09/2011

Hace ya más de un año que abrimos Guía metabólica y nos gustaría conocer tu opinión, tus necesidades y tus propuestas de mejora para el web. También nos interesa saber cómo recursos como este web y otros en Internet están cambiando el papel de los afectados por un ECM y sus familias respecto a su salud.



"El cuidado bueno y cariñoso de los padres es lo más importante que pueden dar a su hijo con un CDG"

19/10/2011

Los Defectos Congénitos de la Glicosilación (CDG)



Questions

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Preliminary evaluation of impact

Changes in daily life and feelings

Discovered new information or increase their knowledge_____93.6%

Changed or developed at least one new habit_____50.1%

Do we offer new
information?

We present
information in plain
language

Preliminary evaluation of impact

Changes in daily life and feelings

Acknowledged that they felt less lonely_____75.4%

Realized that they were doing well when getting in touch with others in the same situation_____72.4%

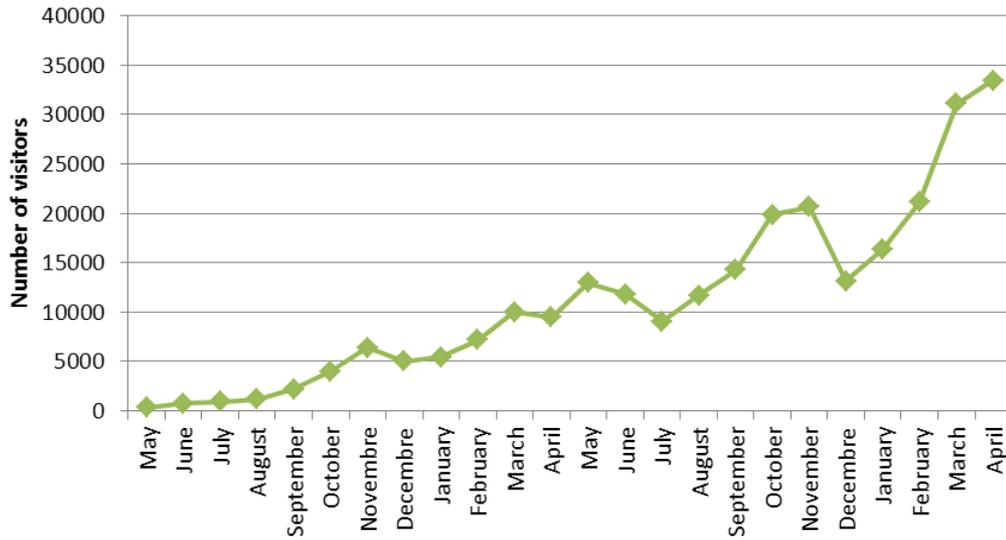
↓ Feelings of loneliness

↑ Self-efficacy

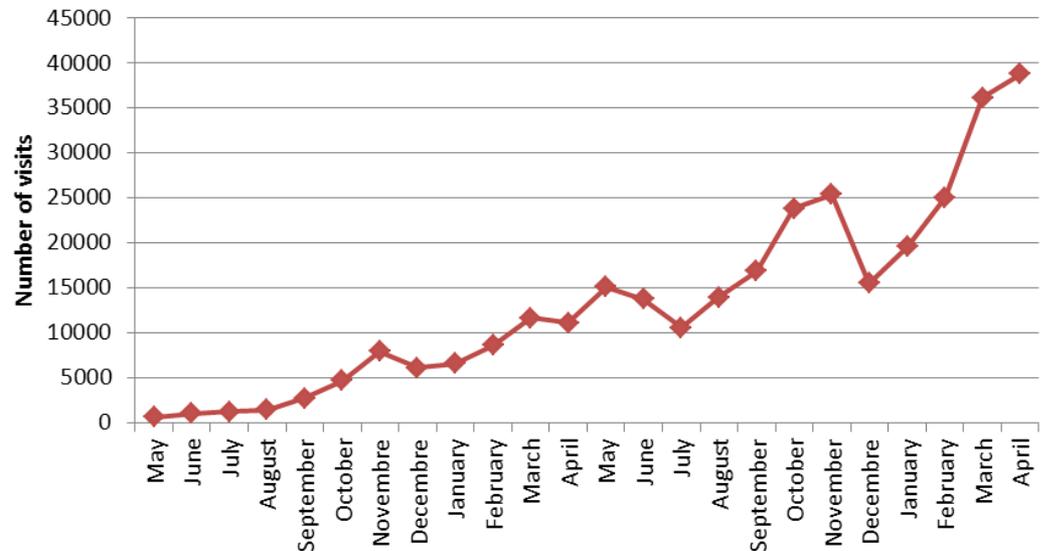
↓ Level of self-criticism

↑↑ Empowerment

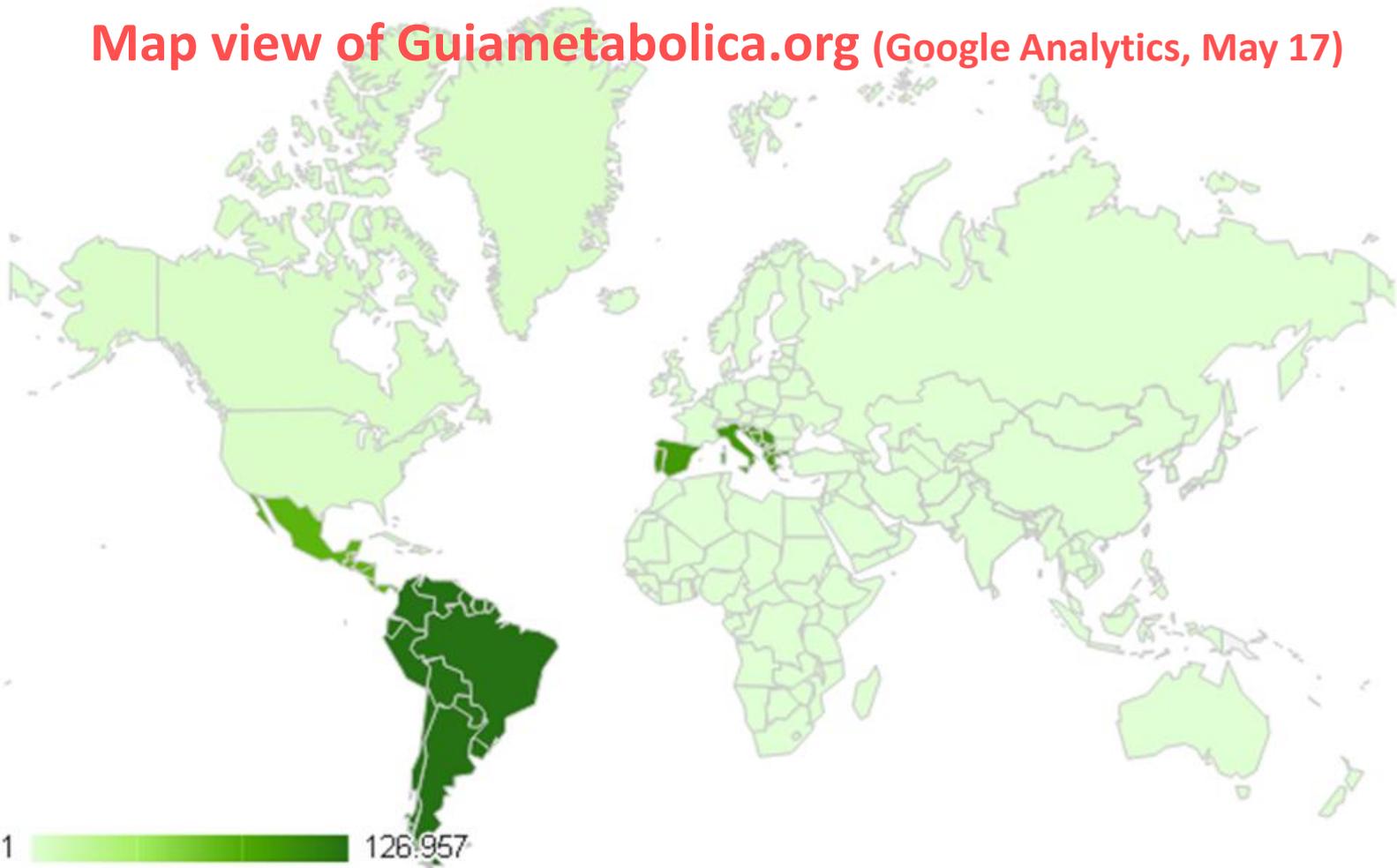
Unique visitors



Visits



Map view of Guiametabolica.org (Google Analytics, May 17)



We register visits from more than **100** countries/territories
More than 70% of the visitors came from different parts of the Western Hemisphere, especially Latin America

Questions

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We have learned/confirmed...

1. Caregivers/patients suffering from RD are one of the most active in the Internet, they constitute the best example of "e-Patients", i. e., health consumers who use the Web 2.0 tools in coping with diseases.
2. Data show that users of Guiametabolica.org are **very participative**.
3. There is a high level of **expertise** in e-caregivers.
4. The **non participative users**, usually called "lurkers" improved their level of knowledge and empowerment in similar degree than most active users.

We have learned/confirmed...

5. Our preliminary data show a decrease in feelings of loneliness among our users, as well as a reduced level of self-criticism.

Taken together, those results probably mean that the web is having an **impact in the psychological and emotional health** of the users.

We have learned that, at least in part, specific websites can offer solutions to some of the **non-considered needs** in the traditional medical care.

We have learned/confirmed...

6. Use of Internet-based health information enhances patients' understanding and their ability to manage their health conditions.

But frequently patients need **supplementary education** on how to assess the quality and relevance of that information.

As a result, physicians have a new role which is often not welcome; furthermore, they are frequently unprepared to handle these patients and often interpret these situations as a threat to their medical expertise.

Perhaps, model needs to be changed: doctors prescribing websites.

Conclusion

Specific IEM websites, especially 2.0 tools and online support groups, should be considered as a possible **complement** to more traditional clinical approaches.

These implements can be offered in the framework of a public care provision and may be an example for those physicians that feel uncomfortable with patients that seek Internet health information or support opening the door to future collaboration between patients and doctors allowing them to have an online direct relationship within in the public health system.

Conclusion

We believe that GuiaMetabolica.org contribution lies in the effect that it has on people's general well being while not interfering with traditional care.



Questions

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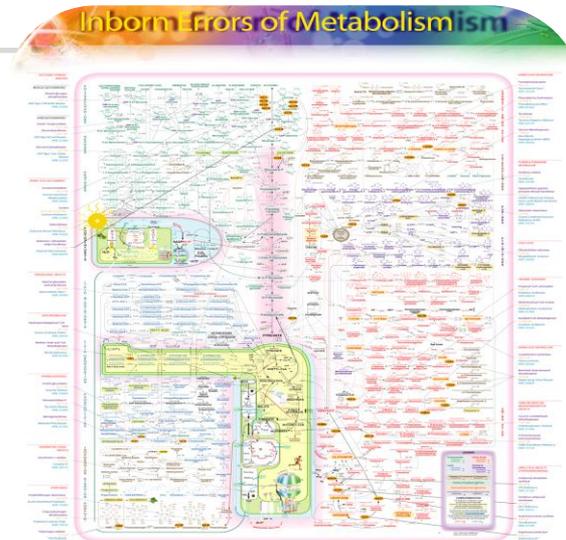
Plans for the future

1. More than 500 Inborn Errors of Metabolism:
58 IEM are included in Guiametabolica.org

2. Increase the target audience:
Translation into English

Guía Metabólica Translation Project

3. Follow offering contents, specific articles,
updated information about research in IEM...



Diseñado por Donald E. Nicholson, The University of Leeds, Inglaterra. <http://www.iubmb-nicholson.org/gif/InbornErrors.html>



american
school OF BARCELONA

guía
m3tabólica

Questions

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Collective Intelligence for Rare Diseases

- **Determinants of Research in RD**

- (1) the human factor and the need to incorporate and train researchers;
- (2) the financial factor, facilitating the sustainability in the long term;
- (3) the access to complete and homogeneous registries;
- (4) and the social factor, including the empowerment of patients and their role as participants in clinical studies or clinical trials.

- **Exploring collaborative models of research**

- (1) participation of patient groups to EU-funded research projects;
- (2) inclusion of patients in research infrastructures
- (3) and patient involvement in each step of clinical trial development.



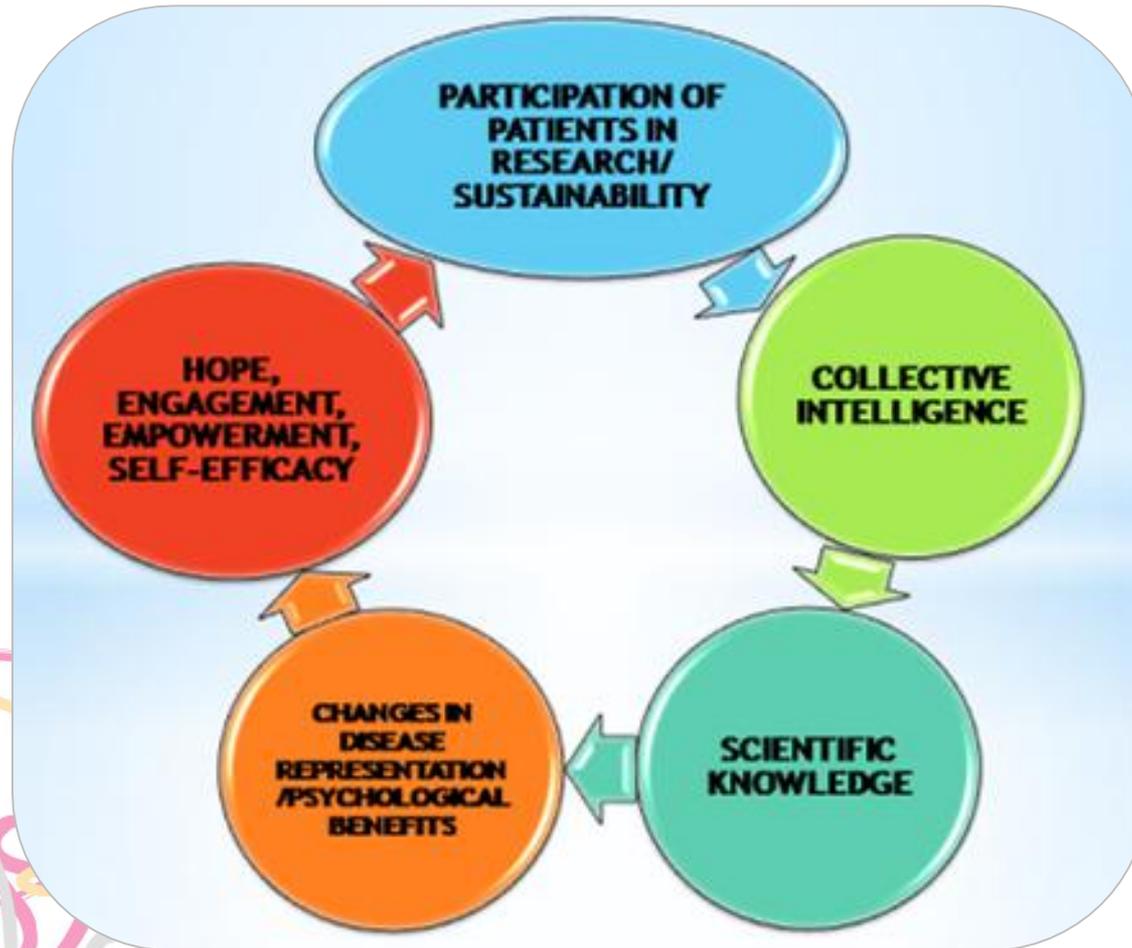
Collective Intelligence for Rare Diseases

Aims

1. To develop a **model** to take advantage of e-Patients potential in several aspects of biomedical research: fundraising, technical collaboration (micro-volunteering, crowd sourcing), direct help to medical research (recruitment, monitoring, etc.).
2. To increase **medical knowledge** on RD (natural history of disease, genotype/phenotype correlations, etc.) through Collective Intelligence: e-patients and researchers collaborating through the Internet.
3. To improve **patients' empowerment**, the perception of self-efficacy and the perceived social support through their direct participation in research.



Collective Intelligence for Rare Diseases



Collective Intelligence for Rare Diseases

- Arrive to those e-patients and e-caregivers that nowadays are using Facebook like social network to share knowledge and experiences about ultraRDs
- A fully-developed prototype for Lowe Syndrome, adaptable to other RDs
- 4 dimensions extracted from patient & research need analysis:
 - (1) Medical knowledge,
 - (2) Psychological /Social aspects,
 - (3) Communication and Dissemination,
 - (4) Fundraising.



<http://www.facebook.com/LoweResearchProject>

Collective Intelligence for Rare Diseases

Expected Results

The virtual platform will have different positive outcomes, both for researchers and for patients/caregivers:



- User generated content: databases generated by patients of RD.
- Closer relationships among patients, researchers, and clinicians: exchange of information, increased reciprocal confidence.
- New sources of knowledge based in the “Wisdom of the Crowd”.
- Positive psychological outcomes between the caregivers: active role vs. passive, empowerment, hope, perceived social support,...

Muchas gracias por su atEnción

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