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,
- Very heterogeneous group
- More than 500 diseases
Addition

1. Lack of information
1. Resources

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

- Patients’ demands & needs
- Real and perceived health status
- Influencing disease
- Provide the services answering the needs of patients in their daily life
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.
We developed a frequently-updated 2.0 website in Spanish for IEM patients.
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

83%

E-CAREGIVERS

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?
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 aminosueros Genes MTHFR

Dietas controladas en Proteínas

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Comentarios

**L CISTINA**
Enviado por majoncfet25 el Lun. 24/05/2012 - 16:12.

DRA. SERRANO
AGRADEZCO INFINITAMENTE SUS RESPUESTAS, MI HIJO VIERTO GRACIAS A DIOS SE ENCUENTRA MUCHO MEJOR, ESTO VINO MUY DELICADO. AL DIA DE HOY, EN CONSTANTE MEJOR.

PODRIA USTED INFORMARME SOBRE EL USO DE LA L CISTINA PARA BAJO EL NIVEL DE HOMOCISTEINA, ES MUY UTILIZO Y AGRADO ZCO LA INFORMACIÓI SALUDOS!

Responder:

**Cieloisa**
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 beneficios.

Sobre todo, volvía 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 déficit de Cbl-C, MTHFR y son diez homocistinas no clínicas.

Adjunto informe sobre el artículo más reciente por si puede resultar de utilidad.


Reciba un cordial saludo,
Dra. M. Serrano
Neurooftalmología. CIBERER. Unidad de Enfermedades Metabólicas

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Suscribirse

Reciba nuestro boletín de noticias
Usuario: HSJD Dra. M. Serrano
DARSE DE BAJA
Boletines anteriores

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Enviar tus recetas
Comment field

Medical/nutritional questions

Share experiences among families

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

- 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
- 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:
Analysis of users’ questions
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?
- **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?
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.
(4) 195 specific recipes for controlled diets (proteins, carbohydrates, fat)
(5) 16 complete menus
Content

Geolocated resources
Content

Games
http://www.fenilcetonuria.es/cocinaPKU.htm
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.
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
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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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.
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.
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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- What was the benefit to families or patients?
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- 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?
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...
Questions

- How platform allowed for interaction between researchers, patients, and families on medical and nutritional questions?
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- **Using social media to stimulate community building between specialists and patients, what tools are needed?**
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