GARD Information Center*

National Center for Advancing Translational Sciences
Office of Rare Diseases Research

National Human Genome Research Institute

U.S. Department of Health and Human Services

NIH National Institutes of Health
Turning Discovery Into Health

DEPARTMENT OF HEALTH & HUMAN SERVICES · USA

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Disclaimer

I have no conflicts of interest with respect to this presentation.

Genetic and Rare Diseases (GARD) Information Center

- The GARD Information Center was established in 2002 in response to language in the Senate Report, 1996; the Senate Report 1999/2000 and P.L. 107-280, (Rare Diseases Act of 2002)
- Funded by the National Center for Advancing Translational Sciences/Office of Rare Diseases Research (NCATS/ORDR) and the National Human Genome Research Institute (NHGRI)/Education and Community Involvement Branch

GARD Services

- Provides free access to accurate, reliable information about genetic and rare diseases to patients and families, healthcare providers, biomedical researchers, and the general public.

- Provides:
  - Generalized curated information on the Web in response to public inquiries.
  - Individualized assistance via phone, e-mail, and online email form.

GARD Staff and Advisors

Specialized information specialist staff

(~7.75 FTE)

- 11 genetic counselors (7 board certified counselors including program managers)
- 1 MD (clinical geneticist) for Spanish or English inquiries

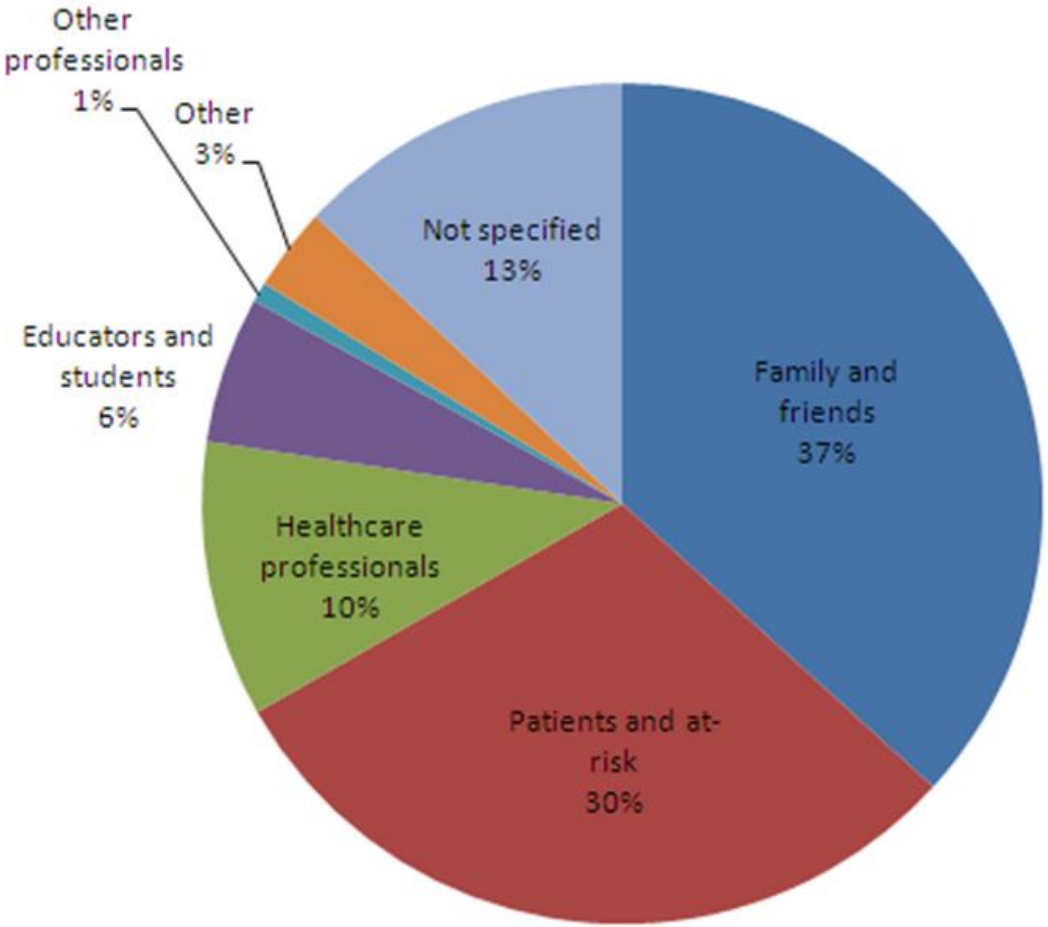
Medical advisors

- Cytogeneticist/molecular geneticist consultant
- Medical consultant for referrals (retired NIH expert)
- Medical geneticist, NIH consultant from NIGMS

Inquiry Statistics

- Answered more than 52,000 questions
- Receive approximately 500 - 600 inquiries/month
- 77% from the United States; 13% International
  - 41% of international inquiries come from the UK, India, and Canada
- 3% of inquiries are in Spanish

Who Contacts GARD?



Frequently Asked Questions

- Where can I find information on symptoms, cause, diagnostic testing, treatment, etc.?
- Are there support groups available?
- How can I get financial assistance?
- Are there research or clinical trials available?
- How can I find an expert?
- How many people have this disease?

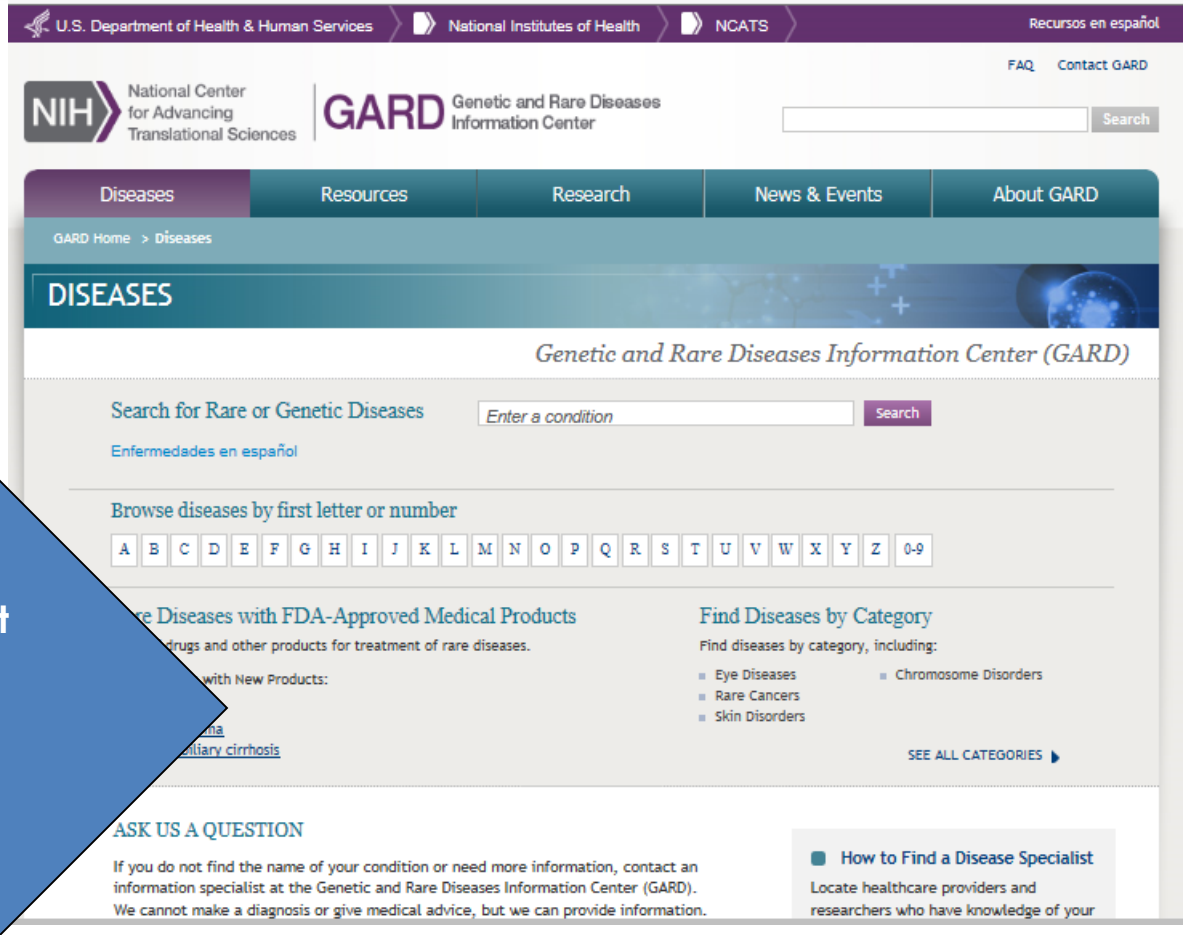
A Sample of Unedited Feedback

"Thank you so very much for your thorough reply to my email. I am so appreciative of your reply. *I was astounded that you replied and nearly brought to tears. It felt like we were not quite as alone as it has felt for too long….*" (Relative)

Many thanks for your incredibly valuable help! I am going to discuss this with my primary care doctor …I know he will be relieved that I am now able to at least attempt to pursue anything that may relieve him of 15 years of scrambling to find referrals…I may not find any answers, but at least I'll have been able to try thanks to you…Again*, many many thanks for taking the time to put together this really great "map of hope", if you will*! (Patient)

GARD Workflow

Web content driven by customized inquiry responses



GARD Website Content

- List of 6,603 rare disease terms (as of 8/31/2015)
  - Most have a Web page for information resources
- More than 2,100 de-identified questions & answers have been posted for 1,400 + diseases
- List of diseases with FDA-approved medical products
- General factsheets (also available in Spanish)
  - Tips for the Undiagnosed
  - FAQs about Chromosome Disorders
  - How to Find a Disease Specialist
- >100 Spanish disease pages with disease-specific information and resources

U.S. Department of Health & Human Services | National Institutes of Health | NCATS | Recursos en español

FAQ | Contact GARD

NIH National Center for Advancing Translational Sciences | GARD Genetic and Rare Diseases Information Center

Search

Diseases | Resources | Research | News & Events | About GARD

GARD Home > Enfermedades En Español

ENFERMEDADES EN ESPAÑOL

Genetic and Rare Diseases Information Center (GARD)

- Search For Diseases
- Browse A-Z
- Disease Categories
- Rare Disease with FDA Approved Medical Products
- Enfermedades en español

Video Sobre GARD

La lista de enfermedades que le proporcionamos a continuación contiene algunas de las condiciones de las cuales GARD ha recibido preguntas en español. Nuestras páginas en la red son constantemente actualizadas de acuerdo a las preguntas recibidas por el Centro de Información sobre Enfermedades Genéticas y Raras (GARD) y siempre que nueva información esté disponible. Aprenda más sobre GARD.

Una condición o enfermedad rara o poco común (o huérfana) es definida como aquella que afecta menos de 200.000 personas en los Estados Unidos. Esta prevalencia es apenas un estimado y puede cambiar en el futuro. El asterisco rojo (*) indica que la condición o enfermedad no es poco común.

Si usted no encuentra información sobre una determinada condición en nuestras páginas de GARD usted puede ponerse en contacto con nosotros.

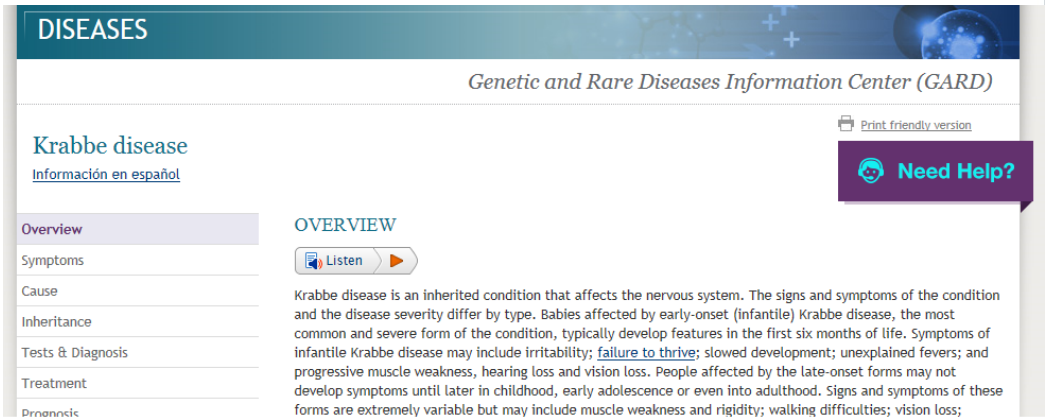
Si usted quiere hacer algunos comentarios sobre la información recibida o enviar sugerencias visite nuestras páginas de GARD sobre comentarios.

* Esta enfermedad no es rara o poca común

Acidemia metilmalónica
Aciduria 2-hidroxiglutárica
Acondroplasia
Adrenoleucodistrofia ligada al X
Agenesia del cuerpo calloso
Amelogénesis imperfecta
Anemia aplásica
* Artritis idiopática juvenil
Asociación VACTERL
Ataxia espinocerebelar
Atrofia muscular espinal
Atrofia muscular espinal tipo 1
Atrofia muscular espinal tipo 2 - Vea Atrofia muscular espinal
Atrofia muscular espinal tipo 3
Atrofia muscular espinal tipo 4 - Vea Atrofia muscular espinal
Bronquiolitis obliterante
Cáncer gástrico difuso
Cáncer gástrico difuso hereditario
Deficiencia de isobutiril-CoA-deshidrogenasa
Deficiencia de la lipasa ácida lisosómica
Deficiencia del transportador de riboflavina
Déficit aislado de hormona de crecimiento
Déficit de la hormona de crecimiento
Disostosis espondilotorácica
Displasia campomélica

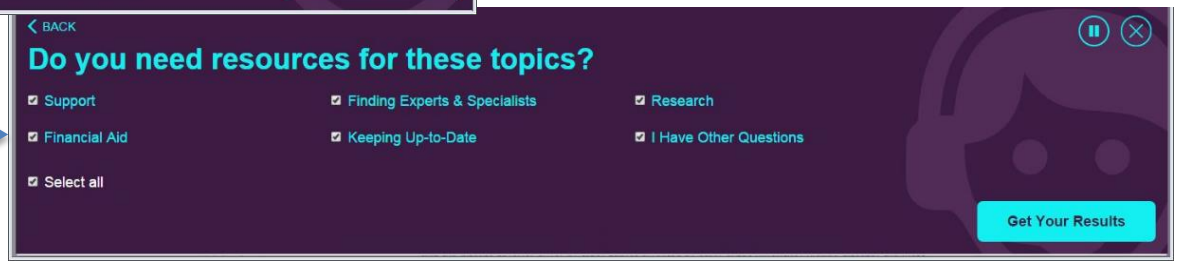
GARD Information Navigator

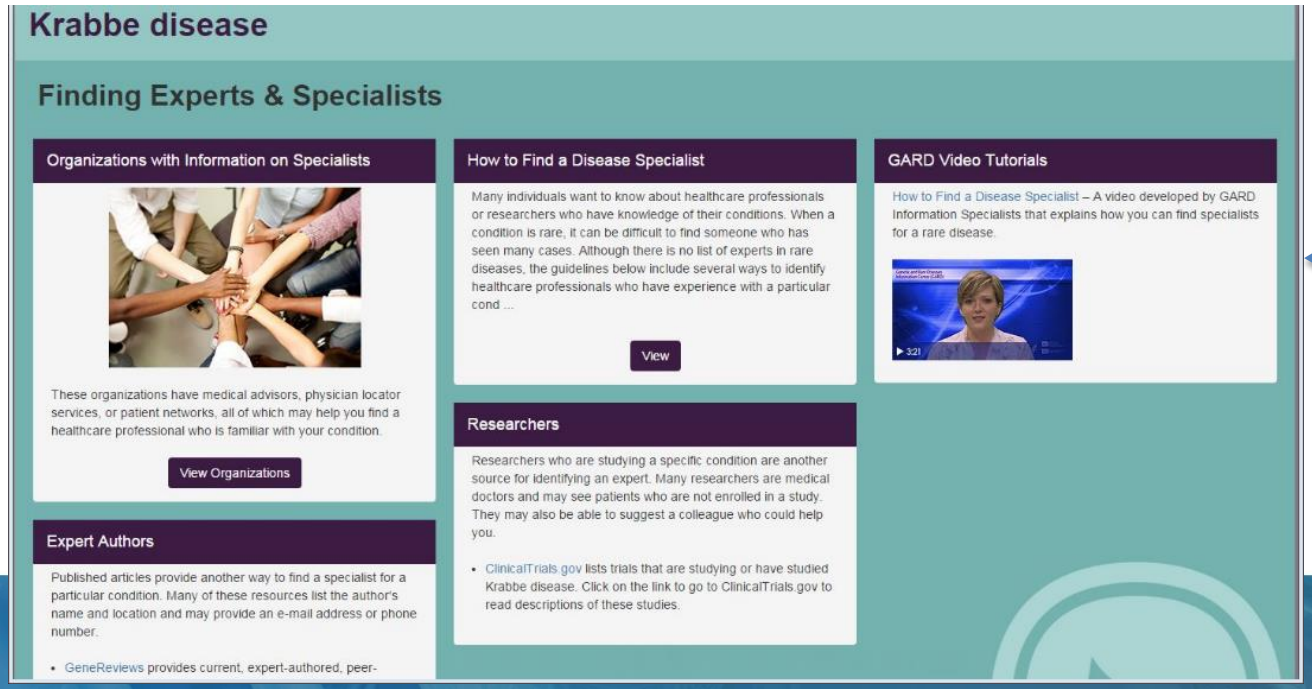
- GARD Information Navigator:
  - launched for 20 diseases on September 2015.



- Audio and interactive navigation to:
  - Aggregate content across the Website
  - Educate users about resources

GARD Information Navigator(cont.)





GARD Website

Statistics

- Approximately 200,000 visits each month
- 42% of visits from mobile devices
- Top 5 pages visited (*in July 2015*)
  - MTHFR gene mutation
  - Undiagnosed Diseases Program
  - Superior Mesenteric Artery Syndrome
  - Hemophagocytic Lymphohistiocytosis

Collaborations

- **Rare disease nomenclature** – collaboration efforts to:
  - map terms to other ontologies to facilitate database interoperability;
  - standardize disease names
  - Active efforts with:
    - National Library of Medicine (NLM)
    - National Cancer Institute/Enterprise Vocabulary Services
    - Online Mendelian Inheritance in Man (OMIM)
    - Orphanet
    - Human Phenotype Ontology/Monarch Initiative
    - International Rare Diseases Research Consortium (IRDiRC), Ontology Working Group
- **Support for Research Efforts**
  - Undiagnosed Diseases Network
  - Centers for Mendelian Genomics (CMG)

Future Directions

- Redesigned GARD Web site
  - Improved search capabilities
- More Web content and images
  - "Glossarized" content
  - Crowdsourcing
- Expanded outreach via social media
  - Facebook
  - Twitter

GARD Contact Information

We welcome your feedback!

Toll-free:	888-205-2311
International:	301-251-4925
TTY:	888-205-3223
Fax:	301-251-4911
E-mail:	GARDinfo@nih.gov
Web site:	http://rarediseases.info.nih.gov/GARD/
Mail:	P.O. Box 8126 Gaithersburg, MD 20898-8126

Thank you for your attention!