



# GARD Information Center\*

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National Center for Advancing Translational Sciences  
Office of Rare Diseases Research

National Human Genome Research Institute

U.S. Department of Health and Human Services



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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**

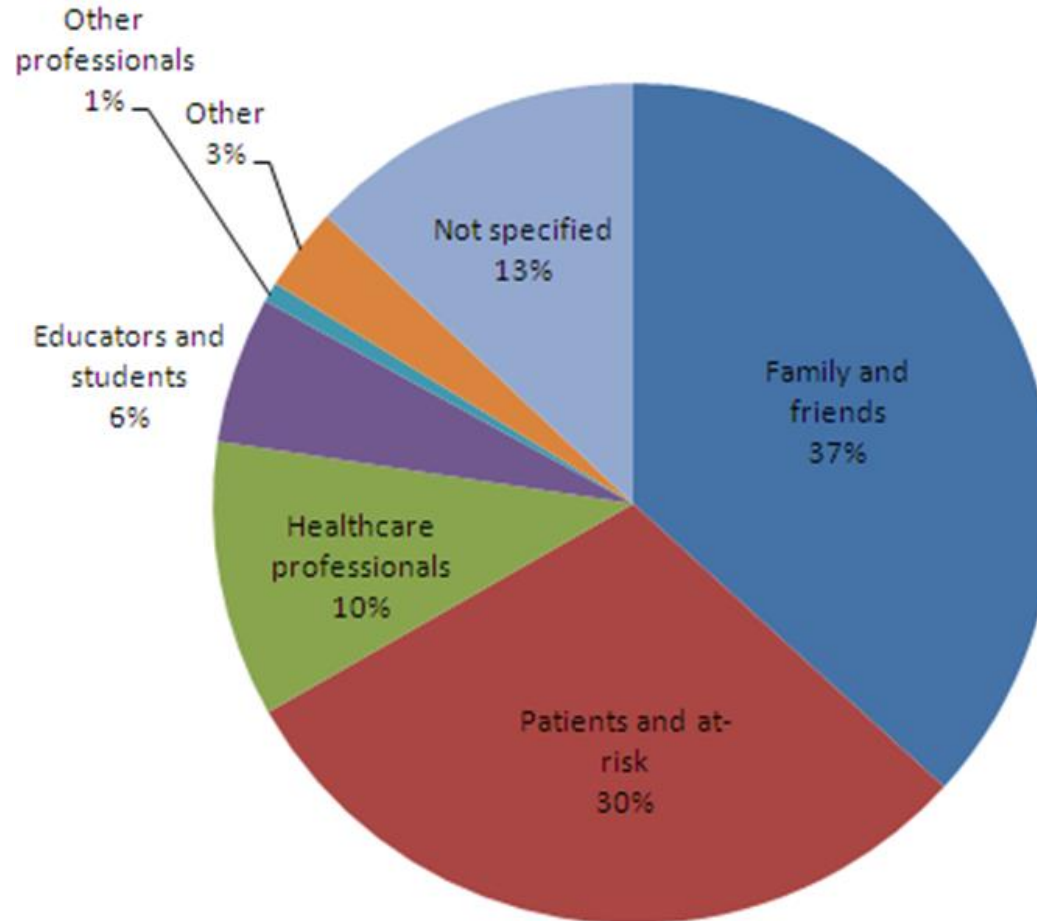
- Cytogeneticist/molecular geneticist consultant
- Medical consultant for referrals (retired NIH expert)
- Medical geneticist, NIH consultant from NIGMS

# 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?



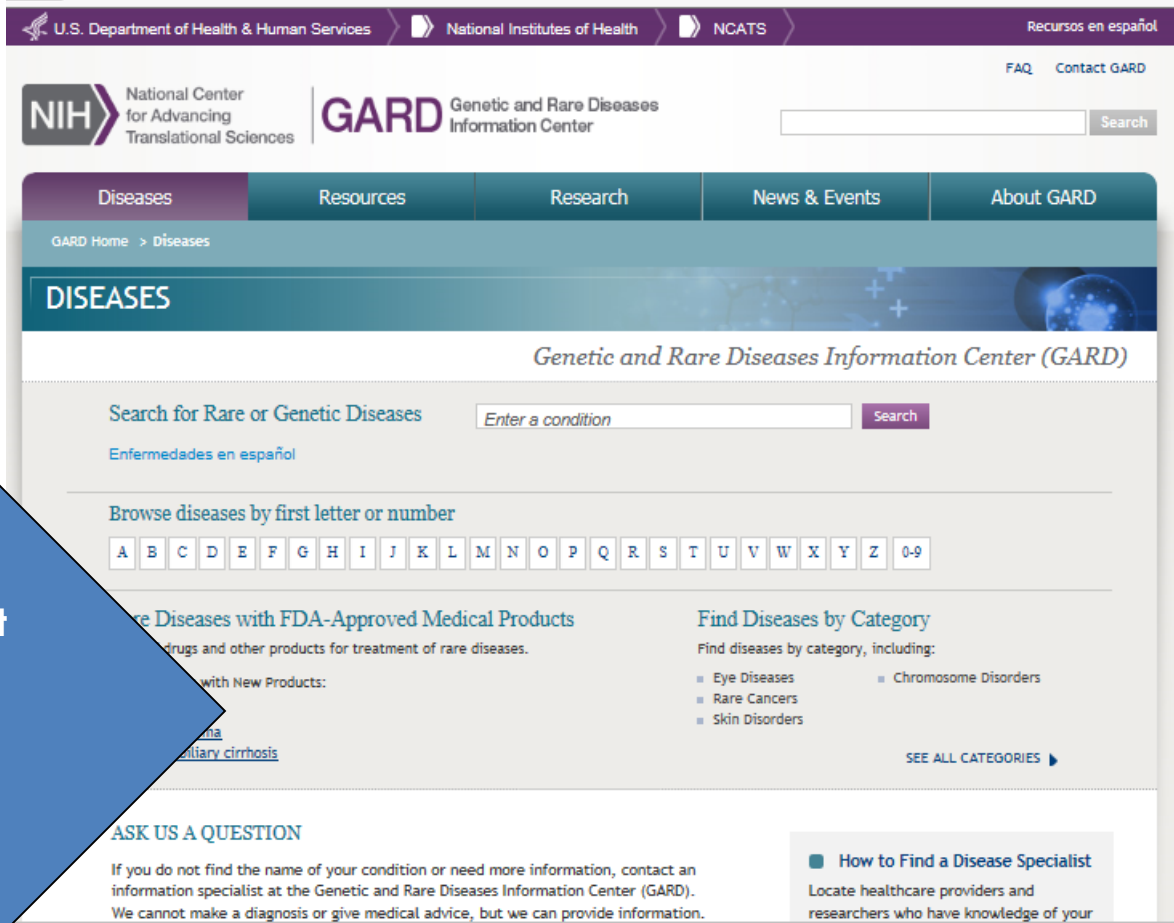
# A Sample of Unedited Feedback



⋮  
“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



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

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

GARD Home > Diseases

## DISEASES

*Genetic and Rare Diseases Information Center (GARD)*

Search for Rare or Genetic Diseases

[Enfermedades en español](#)

Browse diseases by first letter or number

A B C D E F G H I J K L M N O P Q R S T U V W X Y Z 0-9

Rare Diseases with FDA-Approved Medical Products

Drugs and other products for treatment of rare diseases.

with New Products:

[Primary biliary cirrhosis](#)

Find Diseases by Category

Find diseases by category, including:

- Eye Diseases
- Rare Cancers
- Skin Disorders
- Chromosome Disorders

[SEE ALL CATEGORIES](#)

### ASK US A QUESTION

If you do not find the name of your condition or need more information, contact an information specialist at the Genetic and Rare Diseases Information Center (GARD). We cannot make a diagnosis or give medical advice, but we can provide information.

**How to Find a Disease Specialist**

Locate healthcare providers and researchers who have knowledge of your



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

Diseases

Resources

Research

News &amp; Events

About GARD

GARD Home &gt; 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-hidroxiglutarica](#)[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](#)[Distrofia espondilotorácica](#)

# 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

The screenshot shows the GARD Information Navigator interface for Krabbe disease. At the top, a teal header reads 'DISEASES' and 'Genetic and Rare Diseases Information Center (GARD)'. Below this, the title 'Krabbe disease' is displayed with a link to 'Información en español'. A sidebar on the left contains a menu with 'Overview' (highlighted), 'Symptoms', 'Cause', 'Inheritance', 'Tests & Diagnosis', and 'Treatment'. The main content area features an 'OVERVIEW' section with a 'Listen' button (represented by a speaker icon and a play button). The text describes Krabbe disease as an inherited condition affecting the nervous system, noting that signs and symptoms vary by type, with early-onset (infantile) being the most common and severe, typically appearing in the first six months of life. Symptoms include irritability, failure to thrive, slowed development, unexplained fevers, and progressive muscle weakness, hearing loss, and vision loss. Late-onset forms may develop symptoms later in childhood, adolescence, or adulthood. A 'Print friendly version' link is visible in the top right corner, and a 'Need Help?' button is located in the bottom right corner.



# GARD Information Navigator(cont.)

## What would you like to know about Krabbe disease?

- ☐ Signs & Symptoms
- ☐ Cause
- ☐ Inheritance
- ☐ Diagnosis
- ☐ Treatment
- ☐ Statistics
- ☐ Long-Term Outlook
- ☐ Select all

Next: Choose Services

< BACK

## Do you need resources for these topics?

- ☒ Support
- ☒ Finding Experts & Specialists
- ☒ Research
- ☒ Financial Aid
- ☒ Keeping Up-to-Date
- ☒ I Have Other Questions
- ☒ Select all

Get Your Results

## Krabbe disease

### Finding Experts & Specialists

#### Organizations with Information on Specialists



These organizations have medical advisors, physician locator services, or patient networks, all of which may help you find a healthcare professional who is familiar with your condition.

[View Organizations](#)

#### Expert Authors

Published articles provide another way to find a specialist for a particular condition. Many of these resources list the author's name and location and may provide an e-mail address or phone number.

- GeneReviews provides current, expert-authored, peer-

#### How to Find a Disease Specialist

Many individuals want to know about healthcare professionals or researchers who have knowledge of their conditions. When a condition is rare, it can be difficult to find someone who has seen many cases. Although there is no list of experts in rare diseases, the guidelines below include several ways to identify healthcare professionals who have experience with a particular condition ...

[View](#)

#### Researchers

Researchers who are studying a specific condition are another source for identifying an expert. Many researchers are medical doctors and may see patients who are not enrolled in a study. They may also be able to suggest a colleague who could help you.

- [ClinicalTrials.gov](#) lists trials that are studying or have studied Krabbe disease. Click on the link to go to [ClinicalTrials.gov](#) to read descriptions of these studies.

#### GARD Video Tutorials

How to Find a Disease Specialist – A video developed by GARD Information Specialists that explains how you can find specialists for a rare disease.





# 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](mailto: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!