

# Rare Disorders — National Dissemination Center for Children with Disabilities

February 2011

Alone we are rare.  
Together we are strong.”

Slogan of Rare Disease Day  
—February 28, 2011.

Roughly 7,000 rare diseases/disorders have been identified as affecting the human race. Because they are rare, it can be a real challenge for a person to be accurately diagnosed. Finding effective treatments, especially medicine, can also be a challenge—and for the same reason. *Rareness.*

If you have a rare disease, know someone who does, or work with people who might, here’s a core of resources we hope are helpful.



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## Organizations Addressing Rare Disorders

**NORD**, the National Organization for Rare Disorders, at:  
<http://www.rarediseases.org>

**Office of Rare Disorders** at the National Institutes of Health, at:  
<http://rarediseases.info.nih.gov/>

**Genetic and Rare Diseases Information Center (GARD)**  
<http://rarediseases.info.nih.gov/GARD/>

**DiseaseInfoSearch**, for information on specific genetic conditions, at:  
<http://www.geneticalliance.org/>

**Rare Disorders**, at:  
<http://www.rare-disorders.com/>

**The Children’s Rare Disease Network**, at:  
<http://www.crdnetwork.org/>

**The Rare Blog**.  
<http://www.crdnetwork.org/blog/>

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## Genetics and Genetic Disorders

**Three from the Genetic Alliance.** If genetics or genetic disorders interest you, check out these three resources from the Genetic Alliance.

### **Understanding Genetics**

*begins with a basic introduction to genetics concepts, followed by detailed information on topics such as diagnosis of genetic conditions, family history, newborn screening, genetic counseling, understanding patient stories, and ethical, legal, and social issues in genetics.*

<http://www.geneticalliance.org/understanding.genetics>

**Making Sense of Your Genes: A Guide to Genetic Counseling**

*includes general information about genetic counseling, tips on how to prepare for an appointment, and details about different specialties in the field.* <http://www.geneticalliance.org/counseling.guide>

**Students Living With a Genetic Condition: A Guide for Parents**

*You know how to identify and manage symptoms at home, and it may be scary to have someone else manage your child's medical care in your absence. This guide provides information that may be helpful when writing a letter or when preparing to meet with your student's teacher, school nurse, and physical education teacher or coach. This may also be a useful resource for bus drivers, babysitters, church leaders, or any other potential caregiver.* <http://tinyurl.com/23neulr>

**Talking Glossary of Genetic Terms.**

The National Human Genome Research Institute (NHGRI) created the Talking Glossary of Genetic Terms to help everyone understand the terms and concepts used in genetic research. In addition to definitions, specialists in the field of genetics share their descriptions of terms, and many terms include images, animation and links to related terms.

<http://www.genome.gov/Glossary/>

**Frequently asked questions.**

Go to the link below, which is the landing page that can connect you to more info about genetic disorders, genetic testing, genetic counseling and evaluation, and how to locate a genetics professional. There's also information on how genetics professionals help patients interpret and understand genetic information.

<http://www.genome.gov/27527652>

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**The Human Genome Project**

This is one of the most fascinating scientific and research odysseys you'll ever resemble!

**All about the Human Genome Project**

<http://www.genome.gov/10001772>

**What's going on now.**

Visit the National Human Genome Research Institute and see.

<http://www.genome.gov/>

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**Laws You May Not Have Heard of**

**GINA—the law against genetic discrimination.**

With genetic testing becoming increasingly pervasive in medical care and our daily lives, three of the most prominent organizations in genetics (the Genetics and Public Policy Center at Johns Hopkins University, the National Coalition for Health Professional Education in Genetics, and Genetic Alliance) have teamed up to produce educational materials about the Genetic Information Nondiscrimination Act (GINA), a landmark federal law that protects individuals from the misuse of genetic information in health insurance and employment. Start in at:

<http://www.ginahelp.org/>

**U.S. Orphan Drug Act.**

Recognizing that adequate drugs for rare disorders had not been developed in the U.S., and that drug companies would actually incur a financial loss in developing drugs for rare conditions, the U.S. Congress in 1983 passed the Orphan Drug Act. The Orphan Drug Act offers incentives to induce companies to develop drugs (and other medical products) for the small markets of individuals with rare disorders (in the U.S., 47% of rare disorders affect fewer than 25,000 people). Read more at:

<http://www.fda.gov/ForIndustry/DevelopingProductsforRareDiseasesConditions/default.htm>

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## Orphan Drugs

The term “orphan drug” refers to a product that treats a rare disease affecting fewer than 200,000 Americans. The Orphan Drug Act was signed into law on January 4, 1983. Since the Orphan Drug Act passed, over 100 orphan drugs and biological products have been brought to market. ([Stanford University](#), n.d.)

The resources below will help you learn more about orphan drugs—those that have been developed and those currently under development.

### **Video from the FDA.**

*Tim Coté on Orphan Drugs.*

<http://www.fda.gov/AboutFDA/Transparency/Basics/ucm213762.htm>

### **The Office of Orphan Products Development.**

*Where it's happening!*

<http://www.fda.gov/forindustry/developingproductsforrare diseasesconditions/default.htm>

### **Search the Orphan Drug Products Grant Program.**

*What's happening? What's happened?*

<http://www.accessdata.fda.gov/scripts/opdlisting/oopdgrants/>

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## The Undiagnosed Condition

### **Learning about an undiagnosed condition in a child.**

<http://www.genome.gov/17515951>

### **Tips for those with an undiagnosed condition.**

<http://rarediseases.info.nih.gov/GARD/TipsForAnUndiagnosedCondition.aspx>

### **The Undiagnosed Disease Program @ NIH.**

<http://rarediseases.info.nih.gov/Resources.aspx?PageID=31>

### **Mystery Diagnosis | The TV Series.**

Heard of this TV show on Discovery? It frequently deals with rare diseases or disorders that have gone undiagnosed for years. Each episode tells a different person's story—you hear from the person, the medical staff, and the person's family. The link below takes you to the list of diseases the show has highlighted. Maybe your rare disorder is one of them!

<http://health.discovery.com/fansites/mystery-diagnosis/diseases.html>

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**Still can't find any info on the rare disorder that concerns you?** Write to us in English or Spanish at [nichcy@aed.org](mailto:nichcy@aed.org), or give us a call at 1.800.695.0285. We'd be pleased to help.