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Genetics Home Reference: Locating Easy-To-Understand Genetics Information

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CONSUMER HEALTH SEARCHER

Lynda M. Baker, Column Editor

Genetics Home Reference: Locating Easy-to-Understand Genetics Information

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ABSTRACT. This article describes the Genetics Home Reference, the National Library of Medicine's searchable Web resource that provides reliable information about genetic conditions, genes, and chromosomes and their relationship to the health of individuals, families, or populations. This resource contains descriptions of hundreds of genes and genetic conditions, with new content being added regularly. Features of the resource will be described as well as the browse and search methods used to access the information. *[Article copies available for a fee from The Haworth Document Delivery Service: 1-800-HAWORTH. E-mail address: <docdelivery@haworthpress.com> Website: <<http://www.HaworthPress.com>> © 2005 by The Haworth Press, Inc. All rights reserved.]*

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INTRODUCTION

Consumers are increasingly turning to the Internet for health information. According to a national survey conducted by the Pew Internet Project, approximately 6 million Americans go online for medical advice daily.¹ In addition, 93% of health information seekers have gone online to look for information about a particular illness or condition.¹ Given the fact that genetics information is often technical in nature, easy-to-understand genetics information for librarians, consumers, family members, and caregivers is increasingly in demand. Therefore, easily accessible and reliable information in clear language about the relationship between genes and health conditions is needed.

The Genetics Home Reference (GHR) <<http://ghr.nlm.nih.gov>> is an online resource from the National Library of Medicine designed to provide genetics information about the relationship between genetics and health for health educators, librarians, consumers, students, or anyone interested in gaining a better understanding of known gene-condition relationships. This searchable Web-based resource aims to present consumer-oriented information in a clear format and to make the relationship between disease and genetics more understandable. GHR uses clear and concise language, illustrations, and other visual aids to explain the interrelationships between genes and inherited diseases, disorders, or conditions. The resource is freely accessible via the Internet and no registration or personal information is required for use.

ABOUT GENETICS HOME REFERENCE

In April 2003, the National Library of Medicine launched GHR with an extensive range of consumer-oriented information about genetic conditions. Its overall goal is to provide information about genetic conditions, genes, and chromosomes to guide health care consumers in making informed medical decisions. With easy-to-use search and browse capabilities, users can access authoritative summaries on genetic conditions, genes, and chromosomes. A clearly-written and illustrated handbook presents introductory genetics information to enhance the use of GHR. Librarians, health educators, and consumers will ap-

precipitate the colorful visual aids that accommodate various learning styles. Health care consumers can use the Glossary or Search features to find definitions of unfamiliar medical and genetics terms. The glossary provides definitions derived from multiple authoritative sources and in some cases multiple definitions are provided. Consumers may also find the list of synonyms and alternate names provided with each glossary entry useful because medical conditions and genes are often known by multiple terms.

At the time of this review, 179 genetic conditions, 203 gene, and 24 chromosome summaries were available to access, view, and print. In addition, seven *Help Me Understand Genetics* handbook chapters were available. The handbook can be accessed from the main page, from the left column of every condition and gene summary, and also from the Help section on the navigation bar that appears on every page. All GHR content is selected and reviewed by project staff and expert reviewers before release. In addition, GHR content is updated regularly. A disclaimer on the main page explains that information provided should not be used as a substitute for professional medical care or advice. Well-placed links will aid consumers in locating qualified genetics health care professionals in their geographic area.

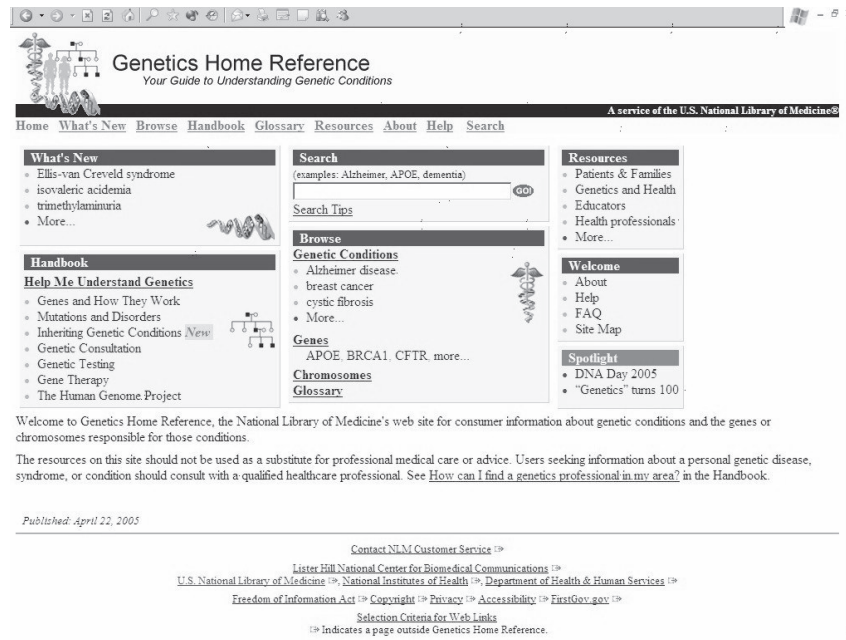
FINDING INFORMATION AT GENETICS HOME REFERENCE

The main Genetics Home Reference page is clear, well organized, and easy to navigate. The Web interface is simple yet visually appealing with many options for searching or browsing (see Figure 1). A navigation bar across the top of every page has menu options that are clearly labeled and allow the user to navigate with ease. The navigation bar includes quick links to the glossary, a Help feature, and the *Help Me Understand Genetics* handbook. The “What’s New” section is prominently displayed on the main GHR page and features recently added content. Content that has been added over the last year is organized by the date of addition allowing easy identification of new topics.

Search and Browse Methods

Health information seekers can easily access genetics information through prominently displayed search and browse features on the main page. A basic search can be performed by entering key words or terms

FIGURE 1. The Genetics Home Reference Web Site



in the search box. The search box is located in the navigation bar. Since the spelling of medical and genetics terms can be challenging, users will value the recommended terms that are presented if misspelled words are entered in the search box. Through the use of Boolean operators (AND, OR, NOT), searches can be refined or multiple search terms combined. Additional assistance with searching techniques is provided by clicking on the “Search Tips” link that is located in the left column of the search results page and is also accessible from the Help section.

Browsing capabilities afford users another method for locating relevant information. GHR content is organized alphabetically as well as by category. Users can browse the alphabetical lists of genetic conditions, genes, or chromosomes. The alphabetical lists are very helpful when users are unsure of the correct spelling. Genetic conditions can also be browsed by categories such as Cancers or Food, Nutrition, and Metabolism. In addition, genes can be browsed by symbol, by full name, or by

categories based on features of their gene products or location of their effects.

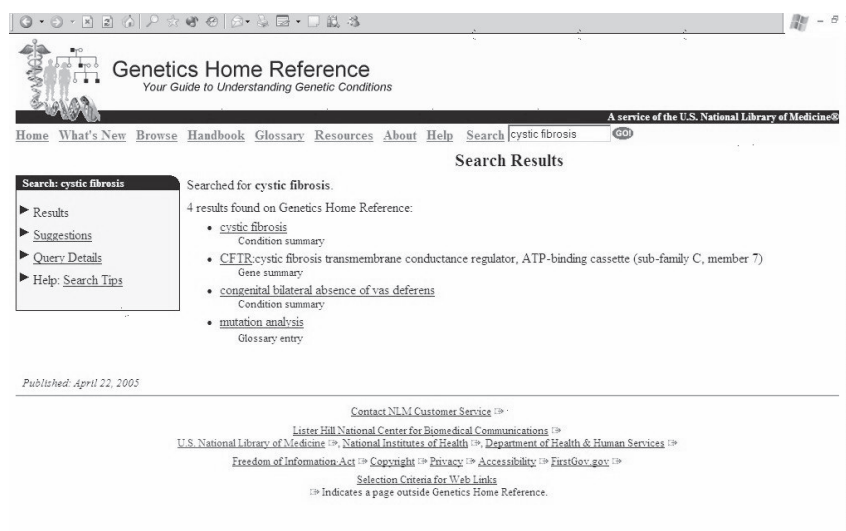
Search Results

The Search Results are displayed in an easy-to-navigate list consisting of genetic condition summaries, gene summaries, and glossary entries. The results are linked to HTML summaries or glossary entries. For instance, a search for “cystic fibrosis” yields content from two condition summaries, a gene summary, and a glossary entry (see Figure 2). The left column of the search results page offers search tips and query details about the search. To access the results, the user simply clicks on the link of interest in the Search Results.

Condition, Gene, and Chromosome Summaries

Summaries are available for genetic conditions, genes, and all human chromosomes. Each summary is presented in a question and answer format and provides basic information on that particular topic with links to additional resources for more information. Hyper-linked tables-of-con-

FIGURE 2. Search Results for “Cystic Fibrosis”



tents appear at the top of every summary enabling users to move easily to any point within a summary. Printer-friendly versions of the summaries are available and related glossary terms are provided at the bottom of each summary. References for resources used in the development of the summaries are provided and lend credibility to the content. In addition, the date of the last comprehensive review of each summary is noted. Health information seekers will find links to relevant support groups and information on genetic testing also contained in the summaries.

Condition Summaries. A condition summary provides basic information about that particular condition, how common it is, genetic causes, and links to additional information such as related genes, patient support groups, and gene tests (see Figure 3). The left column contains links to additional resources that allow the condition summaries to be integrated seamlessly with other related resources from the National Library of Medicine including MedlinePlus <<http://medlineplus.gov>>.

FIGURE 3. Condition Summary for "Cystic Fibrosis"

Genetics Home Reference
Your Guide to Understanding Genetic Conditions

A service of the U.S. National Library of Medicine

Home What's New Browse Handbook Glossary Resources About Help Search

Cystic fibrosis

Print version

Cystic fibrosis

- ▶ **Related Gene(s)**
- ▶ **Quick links to this topic**
 - NIH Publications (3)
 - National Institutes of Health
 - MedlinePlus
 - Health Information
 - Genes and Disease (3)
 - Genetic disorder summaries
 - Educational resources
 - Information pages
 - Patient support
 - For patients and families
 - Gene Reviews (3)
 - Clinical summary
 - Gene Tests (3)
 - DNA test labs
 - ClinicalTrials.gov (3)
 - Research studies
 - PubMed (3)
 - Recent literature
 - OMIM (3)
 - Genetic disorder catalog
 - ▶ **References**

On this page:

- [What is cystic fibrosis?](#)
- [How common is cystic fibrosis?](#)
- [What genes are related to cystic fibrosis?](#)
- [How do people inherit cystic fibrosis?](#)
- [Where can I find additional information about cystic fibrosis?](#)
- [What other names do people use for cystic fibrosis?](#)
- [What if I still have specific questions about cystic fibrosis?](#)
- [What glossary definitions help with understanding cystic fibrosis?](#)

What is cystic fibrosis?

Cystic fibrosis is an inherited disease of the mucus glands. It causes chronic, progressive damage to the respiratory system, chronic digestive system problems, and can affect other organs.

The signs and symptoms of this disorder are caused by the production of abnormally thick, sticky mucus in the body's organs. Problems with breathing are among the most serious symptoms. Mucus can obstruct the airways and cause bacterial infections in the lungs, leading to chronic coughing, wheezing, and inflammation. Over time, mucus buildup and infections lead to permanent lung damage, including the formation of scar tissue (fibrosis) and cysts in the lungs.

Most people with cystic fibrosis also have digestive problems. Mucus can block the ducts of the pancreas, preventing enzymes produced by that organ from reaching the intestines to help digest food. Problems with digestion can lead to diarrhea, malnutrition, and weight loss. Some babies with cystic fibrosis have meconium ileus, a blockage of the intestine that occurs shortly after birth.

Infertility, or the inability to conceive a child, is common in men with cystic fibrosis, but infrequent in women with the condition.

How common is cystic fibrosis?

Cystic fibrosis is a common genetic disease affecting Caucasians (whites) in the United States. The disease occurs in 1 out of every 3,000 Caucasian births. It also affects other ethnic groups, but is much less common: 1 in 15,000 African Americans and 1 in 31,000 Asian Americans have the disorder.

ClinicalTrials.gov <<http://clinicaltrials.gov>>, and PubMed <<http://pubmed.gov>>. Clicking on the link to PubMed gives the user the results of a predetermined search of recent research articles. This is a useful option for those unfamiliar with searching PubMed to identify relevant references to the biomedical literature.

Gene Summaries. A gene summary outlines the normal function of the gene and the conditions associated with mutations in the gene. The official name of the gene is listed and the chromosomal location is illustrated. Links to genetic conditions associated with the gene are provided in the left column of the gene summary, as well as specific information about testing for the gene of interest. Usage reports have shown that the gene summaries are frequently consulted, helping to dispel the notion that information about genes would not be of interest to a consumer audience.²

Chromosome Summaries. Information about each of the 23 pairs of human chromosomes is provided. A chromosome summary follows the question and answer format with a description of the chromosome, conditions caused by changes in the structure or number of copies of the chromosome, and a simple black and white diagram of the chromosome. Links to other resources including National Institutes of Health publications are available for more information. Links to handbook chapters that will assist consumers in gaining a better understanding of chromosomes are also provided.

Additional Features

A useful feature of the Genetics Home Reference is the *Help Me Understand Genetics* handbook. Usage reports have demonstrated that the online handbook is heavily used.² It consists of seven chapters with diagrams and other visual aids to provide basic information about genetics and inherited conditions. The handbook contains easy-to-understand explanations of DNA, genes, proteins, and chromosomes. Handbook chapters also discuss the nature of genetic disorders, how genes control the growth and division of cells, genetic counseling and testing, and gene therapy. The chapter on genetic consultation includes useful descriptions of this health service, what to expect, and how to find a local genetics professional. The chapter on gene testing includes helpful discussions of gene testing, how it is done, and its benefits, risks, and limitations.

CONCLUSION

The Genetics Home Reference is a searchable resource aimed at health information consumers that illustrates how genetics is related to diseases or conditions. The concentration on the relationship between health conditions and genes makes this resource a valuable complement to other consumer health information resources available from the National Library of Medicine, such as MedlinePlus. Value-added features include summaries in clear language, a glossary, an online genetics handbook, and additional links for locating support groups and genetics professionals in a user's geographical area. A "talking" dictionary would be a nice enhancement and could assist health consumers and others to learn the correct pronunciation of challenging technical terms. Overall, the Genetics Home Reference is a unique and extremely valuable resource with flexible searching and browsing capabilities. The additional links to other consumer health information resources as well as to biomedical and research resources offer users pathways to increasingly more in-depth information to support patients and families, educators and genetic scientists. Unlike many consumer health sites, GHR is an easy-to-use resource for locating a wealth of reliable consumer-friendly information about genetic conditions, genes, and chromosomes.

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