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Genetics Home Reference: A Review

Jessica A. Koos
jessica.koos@stonybrook.edu

Ashley Bassett
Rocky Mountain University of Health Professions, abassett@rmuohp.edu

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Jessica A. Koos, Stony Brook University, jessica.koos@stonybrook.edu

Ashley Bassett, Rocky Mountain University of Health Professions, abassett@rmuohp.edu

ABSTRACT. Genetics Home Reference is a free online resource created and maintained by the National Library of Medicine. It is designed to provide genetic information to a wide variety of audiences, particularly the general public. The site consists of original information, as well as links to other curated resources.

KEYWORDS. genetics, consumer health, online databases

AUTHORS.

Jessica A. Koos, M.L.S. (jessica.koos@stonybrook.edu) is a Health Sciences Librarian at Stony Brook University, Stony Brook, NY 11794.

Ashley Bassett, M.L.I.S., M.S. (abassett@rmuohp.edu) is an Assistant Medical Librarian at Rocky Mountain University of Health Professions, Provo, UT 84606.

Introduction

Genetics Home Reference (GHR) is a database provided by the National Library of Medicine that focuses on genetically-related health conditions and their corresponding genes, chromosomes and mitochondrial DNA. It is designed specifically for individuals with little-to-no prior knowledge of genetics, as it is written in plain language that is easy to understand, yet it also provides information that is appropriate for more advanced users. In addition to providing information related to genetic health disorders, there is also a portion of the Web site entitled Help Me Understand Genetics that can aid individuals in understanding the basic components of human genetics and their corresponding functions.

Genetics Home Reference was launched in 2003 in order to make the data obtained from the Human Genome Project more accessible to the public. Prior to the inception of GHR, the resources that were available were not oriented to the needs of consumers, but rather to the scientific community. The creators of GHR sought to bridge that gap by creating a simpler format for complex genetic information, yet links to more detailed information are made available as well.[1]

In 2016, the site underwent a major redesign based on feedback from users, who requested a more updated look.[2] The result is a very clean, modern-looking, easily navigable interface with detailed colored images and photographs displayed throughout the site (see Figure 1).

[PLACE FIGURE 1 HERE]

Text for Figure 1: Figure 1: GHR Home Page

GHR currently contains summaries on over 1,200 health conditions and over 1,400 genes. Users can discover genetic causes, inheritance patterns, signs and symptoms of health

conditions. .In order for a particular condition to be included on the site, specific selection criteria must be met. There must be evidence of a genetic link, and "genetic variations must have been identified in two or more unrelated families." In addition, this information must have been confirmed by studies conducted with human subjects.[3]

The staff of GHR at the National Library of Medicine are responsible for creating the content on the site, and consists of five individuals with advanced health-related degrees.[4] The content is reviewed by international experts in genetics, most of which have either an M.D., Ph.D. or both.[5] In addition, "through a partnership with Genetic Alliance, Genetics Home Reference invites patient support and advocacy groups to provide feedback on Web site content.[6] Some of the information on the site is also automatically extracted from the NCBI Gene and UniProt databases.[7] The information on GHR is also linked to MedLine Plus Connect, a service which connects patients' electronic health records to consumer-friendly health information.[8]

There are also links provided to other Web sites on GHR, and they must meet a list of specific criteria in order to be added to the database. For example, the Web site must provide information that is accurate, up-to-date and information sources must be clearly identified.[3]

Searching the Database

There is a basic search box available on the home page, which will search the entire contents of the site. There are also specific categories that can be browsed as well.

The main search box is located in the upper right-hand corner of the home page. The user can enter any desired search terms and click the magnifying glass icon to proceed. A list of results appears, and the user can either select one of the results, or filter the results by GHR categories (i.e., Health Conditions, Genes, or Help Me Understand Genetics) using the list on the left-hand side of the screen.

There is an option to browse various categories of the database as well. Health Conditions and Genes can be browsed alphabetically by name using the A-Z list. The "Learn More about Health Conditions" box adjacent to the list has links that answer basic questions, such as, "What does it mean if a disorder seems to run in my family?"

Users also may browse through information about each chromosome and mitochondrial DNA using the infographic-style layout on the Chromosomes & mtDNA tab. Clicking on the graphic for a particular chromosome takes users to a page with five collapsible panels with a description of the chromosome, health conditions related to chromosomal changes, a chromosomal diagram, additional resources, and a list of sources for the page.

The Help Me Understand Genetics tab can be browsed as well. There are 13 collapsible panels, each devoted to a different topic. The topics available include Cells and DNA, Mutations and Health, How Genes Work, Gene Families, Inheriting Genetic Conditions, Genetics and Human Traits, Genetic Consultation, Genetic Testing, Newborn Screening, Gene Therapy, The Human Genome Project, Genomic Research, and Precision Medicine. Under each topic, users will find common questions with links to answers to the question, as well as an option to download a printable chapter on the topic in PDF format. For example, under the third topic heading How Genes Work, the first question is "What are proteins and what do they do?" Clicking on the question takes users to a page with the answer--a simple description of proteins and their functions, and a list of common proteins and examples.

Sample Search

A sample search using the term "color blindness" yields multiple results. The preferred terminology for this concept, "color vision deficiency" is the first result in the list (see Figure 2). A preview of the definition of this term is provided under the heading. Selecting this result will

bring you to a page containing the full definition, an image, and accordion-style tabs for Frequency, Genetic Changes, Inheritance Pattern, Diagnosis & Management Procedures, Other Names for this Condition, Additional Information & Resources, and Sources for this Page. Clicking on these tabs uncovers the information for each heading.

[PLACE FIGURE 2 HERE]

Text for Figure 2: Figure 2: Sample Search

Additional Resources

Genetics Home Reference also provides a Resources tab that includes links to reliable online genetics resources. The links are organized under sixteen genetics-related topic headings, and they include Support and Advocacy, Financial Assistance, General Genetics, Genetic Testing, Disease Registries, Classroom Resources, Clinical/Professional Resources, Genetics Research, Research Participation, Bioinformatics Databases, Genetics News, Policy and Ethics, Glossaries and Medical Terms, Finding and Understanding Medical Journal Articles, Evaluating Health Information on the Web, and Resources for Science Writers. Under each dropdown tab heading, users will find 10-15 links related to the topic heading.

The home page of Genetics Home Reference also features a New & Updated Pages section, where users can see lists of new pages and recently updated pages. At the bottom of each newly updated page, users can see when the page was reviewed and updated. For example, the Leprosy page was reviewed in February 2018, and updated on February 20, 2018. The Bulletins section lists important dates and events that are relevant to genetics, such as Rare Disease Day on February 28, 2018. The Featured section highlights relevant questions such as "What is precision medicine?" GHR also has a Share this Page option in the top right hand corner, which offers options for sharing the page via Facebook, Twitter, or email.

Near the bottom of the GHR home page, users will find a Data Files & API link. This provides information about the file formats available for download for search results and topics on the Web site. The formats provided are XML and JSON.

Conclusion

When compared with other NIH databases, Genetics Home Reference is simpler and more user-friendly. As mentioned previously, GHR is meant to be lay-friendly, making it a unique health information resource. While NIH databases such as Gene offer advanced search options and use expert terminology, GHR features a simple search and lists each term in the results with a definition.

Users will find this Web site useful for genetics-related health questions, and genetics research. Librarians will find GHR helpful when faced with a genetics reference question, and can feel comfortable sending patrons to GHR, thanks to its consumer-friendly layout and language. Health professionals will also find GHR useful for their own genetic information needs, as well as for the needs of their patients.

For More Information

For additional information on Genetics Home Reference, please contact:

The National Library of Medicine

8600 Rockville Pike

Bethesda, MD 20894

(888)346-3656

There is also a “Customer Support” link on the bottom of each Web page on the GHR Web site, which leads to NLM’s Customer Support Web site.

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