Case Report

Community Genetics

Community Genet 2006;9:274–278
DOI: 10.1159/000094477

'Genetics Home Reference': Helping Patients Understand the Role of Genetics in Health and Disease

Cathy Fomous[a] Joyce A. Mitchell[b] Alexa McCray[a, c]

[a]National Library of Medicine, Bethesda, Md., [b]School of Medicine, University of Utah, Salt Lake City, Utah, and [c]Medical School, Harvard University, Boston, Mass., USA

Key Words
Consumer information, genetics · Chromosomal disorders · Medical informatics applications

Abstract
The surge of information generated by the Human Genome Project has left many health professionals and their patients struggling to understand the role of genetics in health and disease. To aid the lay public and health professionals, the US National Library of Medicine developed an online resource called 'Genetics Home Reference' (GHR), located at http://ghr.nlm.nih.gov/. Launched in April 2003, GHR's goal is to help the public interpret the health implications of the Human Genome Project. It bridges the clinical questions of consumers and the rich technical data emerging from the sequenced human genome. The GHR web site is designed for easy navigation among summaries for genetic conditions and the related gene(s) and chromosome(s). This design strategy enhances the user's appreciation of how genes, chromosomes, and conditions are interrelated.

Introduction

The Human Genome Project amplified interest in genetics and propelled medicine into a new era in which genetic knowledge will help determine optimal health care [1, 2]. The surge of genetic information generated by the Human Genome Project is overwhelming, however, and often leaves patients and health professionals struggling to understand the implications of genetic findings in medical practice [1, 3]. Several organizations, such as the World Health Organization and the International Society of Bioethics, recognize that the goals of medical genetics can be fulfilled only in the context of an educated and informed public [4–6]. The healthcare community is challenged to communicate the complex developments in human genetics in a way that the public can freely access, easily understand, and appropriately apply [7].

Increasingly, the public is seeking health information online [8, 9], including information about inherited disorders [10, 11]. Consumers, however, report that genetics web sites are often confusing, difficult to navigate, and hard to understand [7, 12]. To address this confusion, the US National Library of Medicine developed an online resource called the Genetics Home Reference (GHR), located at http://ghr.nlm.nih.gov/.

KARGER

Fax +41 61 306 12 34
E-Mail karger@karger.ch
www.karger.com

1422–2795/06/0094–0274$23.50/0

Accessible online at:
www.karger.com/cmg

Cathy M. Fomous
National Library of Medicine
8600 Rockville Pike, Bldg. 38A, Room 7S704
Bethesda, MD 20894 (USA)
Tel. +1 301 496 0433, Fax +1 301 402 0118, E-Mail fomous@nlm.nih.gov

Goal and General Overview

Launched in April 2003, GHR's goal is to help the public interpret the health implications of the Human Genome Project. It bridges the clinical questions of consumers and the rich technical data emerging from the sequenced human genome. Using a question and answer format, GHR describes genetic conditions and the gene or chromosome alterations associated with those conditions. Information for more than 160 genetic conditions and 250 genes is currently available. New topics are added continuously, and the information is monitored regularly to ensure that it is accurate and up-to-date. Experts in clinical genetics review each topic before it is posted to the GHR site and annually thereafter (for details see http://ghr.nlm.nih.gov/ghr/ExpertReviewers).

GHR's target audience is members of the general public who are motivated to learn about genetic conditions. GHR, however, also serves as an educational site for students and journalists. For health professionals, the site provides information suitable for patients as well as links to genetic resources with primary data and detailed clinical and technical discussions. By integrating a spectrum of resources, from patient-friendly fact sheets to technical databases, GHR serves the public, academic, clinical, and research communities.

General Design

The GHR web site is designed for easy navigation among summaries for genetic conditions and the related gene(s) and chromosome(s) [13]. This design strategy enhances the user's appreciation of how genes, chromosomes, and conditions are interrelated. Using muscular dystrophy (Duchenne and Becker types) as an example, users can navigate from the condition summary for muscular dystrophy (fig. 1a) to the DMD gene (fig. 1b) and then to a summary for the corresponding X chromosome (fig. 1c). From the summary about the X chromosome, there is a link to related chromosomal disorders such as Turner syndrome (fig. 1c). Navigation is not unidirectional. Users can begin with a gene or chromosome summary and follow the links to related conditions.

The GHR design also accommodates four possible relationships between genetic alterations (on a gene or chromosome level) and particular conditions. These relationships are: (1) causal, as HBB mutations cause sickle cell anemia or trisomy 21 causes Down syndrome; (2) predisposing, as a particular COL1A1 polymorphism appears to increase the risk of developing osteoporosis; (3) associated, as TP53 mutations or chromosome translocations are associated with certain cancers, and (4) modifying, as HFE mutations modify the course of X-linked sideroblastic anemia. A particular gene may have more than one relationship, such as FGFR3 mutations in the germ line can cause achondroplasia, but FGFR3 mutations in somatic cells may increase the risk for bladder cancer.

Content

The content for each topic summary is derived from the primary literature and other sources such as US federal agencies, academic sites, and professional organizations. Each summary includes a bibliography from the link to references.

Medical and genetics guidelines [14, 15] are used to name conditions, and alternate names are listed as synonyms. Each condition summary provides a concise description of the disorder and an explanation of incidence or prevalence, inheritance pattern, and related genetic factors such as gene mutations or chromosome alterations. Condition summaries link to a range of educational and patient support resources such as MedlinePlus [16], ClinicalTrials.gov [17] and GeneTests [18].

Gene summaries use the official gene name and symbol, as determined by the HUGO Gene Nomenclature Committee [19]. Each summary explains the gene's normal function and how mutations in the gene cause, modify, or are otherwise associated with particular genetic conditions. Gene summaries link to resources such as Entrez Gene [20] and GeneTests [18].

Chromosome summaries provide an estimate of the number of genes on each chromosome, and some include a discussion of related chromosomal disorders. These summaries link to chromosome-specific resources such as MapViewer [21], Ensembl [22], Chromosome Launchpad [23], and other chromosome databases.

Learning Aids

Recognizing that the target audience may have a limited science background, GHR offers tools to help the motivated learner. Each web page links to a glossary of genetic and medical terms. In addition, each summary provides a list of glossary terms used on the page, with a direct link to their definitions. The GHR glossary is built from the following established sources: the Unified Med-

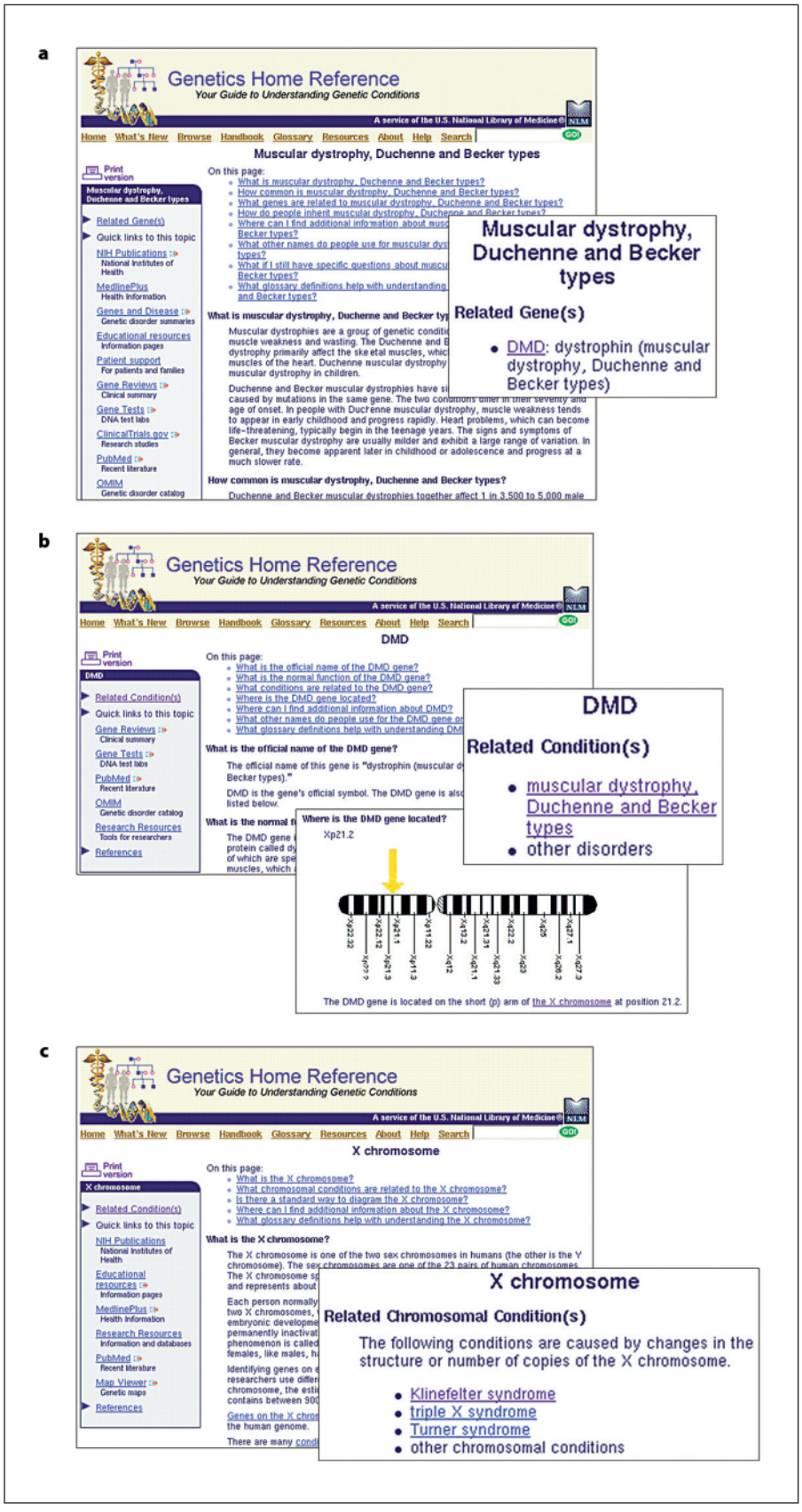

Fig. 1. GHR is designed to ease navigation among summaries for genetic conditions and the related gene(s) and chromosome(s). **a** The summary for muscular dystrophy, Duchenne and Becker types, links to the related gene, DMD. **b** The DMD gene summary provides links to related conditions and also to the X chromosome, where the DMD gene is located. **c** The X chromosome summary links to chromosomal conditions such as Turner syndrome.

ical Language System (UMLS) [24], GeneTests [18], the US Department of Energy Human Genome Project [25], and three components of the US National Institutes of Health [26–28].

An online tutorial is available through a feature called the 'Help Me Understand Genetics Handbook', an illustrated primer that explains the basics of genetics. The handbook is organized by chapters that provide information about how genes work, types of gene mutations, patterns of inheritance, the role of a genetics professional, genetic testing, gene therapy, pharmacogenomics, and the Human Genome Project.

Additional Resources

The GHR site also serves as a conduit to many additional online resources from the US and other countries. Links to condition-, gene-, or chromosome-specific information on other web sites are available from each summary page. Additionally, from the 'Resources' link in the tool bar on each web page, GHR users will find links to genetics news, policies and ethics, online exhibits, the Human Genome Project, genetics and health, and educational tools. GHR makes available its criteria in selecting web sites as additional resources. Briefly, these sites should be educational and free of advertisement for commercial products. There should be no charge or registration required to use the site. The source of the site's content should be clearly visible, respected, and dependable, and the information should be current.

Evaluation and Usage

Preliminary surveys, conducted in early 2003, helped to assess GHR's layout, navigational design, and level of content. Feedback from these surveys influenced the question-answer format of summaries, glossary content, Handbook chapters, and the types and placement of links to additional resources. In early 2004, a formal survey was conducted among members of the Genetic Alliance [29], an international coalition that represents individuals with genetic conditions. The survey provided feedback about GHR's design and content. The results [30], based on 374 respondents, indicated overall user satisfaction – 88% of users were satisfied or very satisfied with the site. Respondents found GHR to be authoritative, accurate, unbiased, up-to-date, and informative.

Since its launch two years ago, GHR site traffic has increased nearly 10-fold. The median daily use based on unique Internet protocol (IP) addresses rose from 466 in May 2003 to 4,213 in May 2005. Tracking data show that the most popular features of the site are summaries of genetic conditions, the genetics tutorial handbook, and the glossary.

Summary and Future Plans

GHR fills a unique niche by using plain language to interpret the health implications of the Human Genome Project. The site provides consumer-friendly information about genetic conditions and the genes and/or chromosomes that play a role in those conditions, along with a range of resources – from basic tutorials to web sites with clinical and technical information. Long-range plans include addressing multifactorial chronic conditions, epigenetics, and the role of single nucleotide polymorphisms. Additional evaluations among motivated health-seeking Internet consumers will help assess GHR's usability and its content readability and impact. GHR will grow and evolve to help consumers navigate the complex world of genetics.

Acknowledgments

We would like to thank the other members of the Genetics Home Reference team: M. Cheh, S.L.H. Davenport, E. Dorfman, J. Fun, J. Gillen, N. Ide, R. Loane, R. Logan, S.M. Morrison, D.M. Mucci, P. Wolfe and K. Zeng as well as all the expert reviewers. We extend special thanks to J. Fun for assisting with the images in this paper.

References

1 Guttmacher AE, Collins FS: Genomic medicine – a primer. N Engl J Med 2002;347:1512–1527.
2 Guttmacher AE, Collins FS: Welcome to the genomic era. N Engl J Med 2003;349:996–998.
3 Khoury MJ: Genetics and genomics in practice: the continuum from genetic disease to genetic information in health and disease. Genet Med 2003;5:261–268.
4 World Health Organization: Review of Ethical Issues in Medical Genetics; Section 3.1. Geneva, WHO, 2003. http://www.who.int/genomics/publications/en/ethical_issues_in_medgenetics%20report.pdf.
5 World Health Organization: Report of a WHO Meeting on Collabortion in Medical Genetics; Section 3.4. Geneva, WHO, 2002. http://whqlibdoc.who.int/hq/2002/who_HGN_WG_02.2.pdf.
6 International Society of Bioethics: Bioethics Declaration of Gijon 2000. Gijon, 2000. http://www.sibi.org/ingles/declaracion.htm.
7 Bernhardt JM: Perceived barriers to Internet-based health communication on human genetics. J Health Commun 2002;7:325–340.
8 Cline RJ, Haynes K: Consumer health information seeking on the Internet: the state of the art. Health Educ Res 2001;16:671–692.
9 Fox S, Fallows D: Internet Health Resources 2003. Washington, Pew Foundation, 2003.
10 Guttmacher AE: Human genetics on the web. Annu Rev Genomics Hum Genet 2001;2:213–233.
11 Taylor MR, Alman A, Manchester DK: Use of the Internet by patients and their families to obtain genetics-related information. Mayo Clinic Proc 2001;76:772–776.
12 Mitchell J, McCray A, Bodenreider O: From phenotype to genotype: issues in navigating the available information resources. Methods Inf Med 2003;42:557–563.
13 Mitchell JA, Fun J, McCray AT: Design of genetics home reference: a new NLM consumer health resource. JAMIA DOI: 10.1197/jamia.M1549.
14 McKusick VA: Preface; in McKusick VA: Mendelian Inheritance in Man: A Catalog of Human Genes and Genetic Disorders, ed 12. Baltimore, Johns Hopkins University Press, 1998, pp xlviii–liii.
15 Smith JM: Eponyms; in Flanagin A, Phil B, Fontanarosa M, et al. (eds): American Medical Association Manual of Style: A Guide for Authors and Editors, ed 9. Baltimore, Williams & Wilkins, 1998, 9th edition, pp 469–471.
16 US National Institutes of Health, National Library of Medicine: MedlinePlus. http://medlineplus.gov/ (August 2004).
17 US National Institutes of Health, National Library of Medicine: ClinicalTrails.gov. http://clinicaltrials.gov/ (August 2004).
18 University of Washington, Seattle: GeneTests. http://www.genetests.org/ (August 2004).
19 University College, London: HUGO Genome Nomenclature Committee (HGNC). http://www.gene.ucl.ac.uk/nomenclature/ (August 2004).
20 US National Institutes of Health, National Library of Medicine, National Center for Biotechnology Information: Entrez Gene. http://www.ncbi.nih.gov/entrez/query.fcgi?db = gene (June 2005).
21 US National Institutes of Health, National Library of Medicine, National Center for Biotechnology Information: Map Viewer. http://www.ncbi.nlm.nih.gov/mapview/ (August 2004).
22 The Wellcome Trust and Sanger Institute: Ensembl Human MapView. http://www.ensembl.org/Homo_sapiens/mapview/ (August 2004).
23 US Department of Energy, Human Genome Project Information: Chromosome Launchpad. http://www.ornl.gov/sci/techresources/Human_Genome/launchpad/ (August 2004).
24 US National Institutes of Health, National Library of Medicine: Unified medical language system.http://www.nlm.nih.gov/research/umls/ (August 2004).
25 US Department of Energy, Human Genome Project Information: Genome glossary. http://www.ornl.gov/sci/techresources/Human_Genome/glossary/ (August 2004).
26 US National Institutes of Health, National Cancer Institute. Dictionary of Cancer Terms: http://cancer.gov/dictionary/ (August 2004).
27 US National Institutes of Health, National Human Genome Research Institute: Talking glossary of genetic terms. http://www.genome.gov/glossary.cfm/ (August 2004).
28 US National Institutes of Health, Office of Rare Diseases: Terms and definitions. http://ord.aspensys.com/asp/resources/glossary_a-e.asp/ (August 2004).
29 http://www.geneticalliance.org/ (June 2005).
30 Peng Z, Logan RA. Content quality, usability, affective evaluation, and overall satisfaction of online health information. Presented to Health Communication Division, International Communication Association Annual Meeting. May 2005.