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Genetics Home Reference;

A Report to the Board of Scientific Counselors

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EXECUTIVE SUMMARY

Genetics Home Reference (GHR) is a credible, comprehensive, and dynamic web site that uses lay language to explain the effects of genetic variation on human health. The site's design allows users to navigate the complex interrelationships among conditions, genes, and chromosomes. In addition, the site provides multiple resources for a broad range of users with varied educational backgrounds. For example, GHR links to research and clinical databases, designed for genetics professionals and also offers learning aids for the genetic novice, such as glossary definitions and tutorials from the *Help Me Understand Genetics Handbook*. The GHR project supports many of the goals of National Library of Medicine's (NLM) long range plan, particularly by providing "access to health information that is useful both to the general public and to practitioners who need information outside their particular field of expertise."

Lister Hill National Center for Biomedical Communications (LHNCBC) offers an ideal setting to address consumer education in genetics. The innovative techniques developed through informatics research at LHNCBC are key to the effective management of a large and expanding body of genetics information. Many of GHR's strategies are based on mining information from existing research, clinical, and consumer databases such as Entrez Gene and MedlinePlus. In addition to using references inherent in these databases, GHR uses semantic information from the Unified Medical Language System to help join relevant information across multiple sources. Semantic tools such as searching with expanded synonymy, indexing to NLM's Medical Subject Heading (MeSH) terms, and extending searches using MeSH relationships help join records describing similar concepts. GHR is a strategic site for investigating how informatics techniques can help in the development and retrieval of health information for the lay audience.

Feedback from formal and informal evaluations have helped shape GHR's layout, navigational design, and level of content. A formal survey of 374 Genetic Alliance members found GHR to be authoritative, accurate, unbiased, pertinent, up-to-date, and informative. The three different prisms by which survey respondents perceive GHR indicate that online users have significant perceptual differences about the web site's image, even when they agree overall on the positive utility of the site. Evaluations of how audiences assess a healthcare web site's image may provide fresh insights about GHR and other health information web sites.

Since its launch two years ago, GHR has displayed steady growth in content and number of visitors. As GHR continues to improve and expand, it will explore more challenging research problems. Continued development of the site will guide the use of informatics techniques and drive new research to assist in selecting new topics, developing content, ensuring accuracy and currency, and helping consumers navigate the complex world of genetics.

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BACKGROUND AND SIGNIFICANCE

Genetics Home Reference (GHR) is a web-based resource for consumers that provides information about genetic conditions and the gene or chromosome variations that contribute to those conditions. This web site fills a unique niche by using lay language to interpret the health implications of the Human Genome Project; GHR is also unique in traversing the complete spectrum of information from consumer questions to the details of gene function and sequences. Prior to the launch of GHR in April 2003, online genetic resources, such as GeneTests [1] and Entrez Gene [2], focused on the needs of genetics professionals. The content of these sites, laden with clinical and technical terms, is often difficult for the general public to understand [3].

The Human Genome Project amplified interest in genetics and is propelling medicine into a new era in which genetic knowledge will contribute to optimal health care [4, 5]. The surge of genetic information generated by the Human Genome Project can be overwhelming and often leaves the public struggling to understand the role of genetics in their health care [4, 6].

Increasingly, the public is seeking health information online [7, 8], including information about inherited disorders [9, 10]. Consumers, however, report that genetics web sites are often confusing, difficult to navigate, and hard to understand [3, 11]. 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 [11].

The Lister Hill National Center for Biomedical Communications (LHNCBC) offers an ideal setting to address consumer education in genetics. GHR complements other consumer health resources at the National Library of Medicine (NLM), such as MedlinePlus [12]. The innovative techniques developed through informatics research at LHNCBC are keys to the effective management of a large and expanding body of genetics information. Also, the infrastructure used to support other web sites, such as ClinicalTrials.gov [13], can be leveraged to support GHR.

PROJECT OBJECTIVE AND SUPPORT OF NLM'S LONG RANGE PLAN

The objective of GHR 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. In addition to a description of selected genetic disorders, GHR uniquely provides an explanation of the normal function of related genes and discusses the effects of gene alterations.

Information about more than 130 genetic conditions and 220 genes is currently available (see Table 1). The GHR staff's goal is to add four new topics to the public site each month; this goal was exceeded in 2004 (see Figure 1). The content is monitored regularly to ensure that it is accurate and up-to-date. Experts in genetics review each topic before it is posted to the GHR site and annually thereafter.

Recognizing that a lay audience may have a limited science background, GHR offers tools to help the motivated learner. Each condition, gene, and chromosome summary provides a list of glossary terms used on the page, with a direct link to their definitions. In addition, a link to a searchable glossary of genetic and medical terms appears on all web pages. An online tutorial is also available through a feature called the *Help Me Understand Genetics Handbook*, which explains the basics of genetics. This illustrated *Handbook* provides 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. The *Handbook* is a dynamic document, and new topics and illustrations are added as needed to support content in the condition, gene, and chromosome summaries.

The GHR project supports NLM's long range plan [14], particularly priorities related to health information for the public. NLM recognizes that it should "provide access to health information that is useful both to the general public and to practitioners who need information outside their particular field of expertise." GHR supports goals within the long range plan by organizing authoritative biomedical information for the general public and by using feedback to improve the site. GHR staff members participate in outreach activities to promote awareness of the site among health professionals and the public. The staff is collaborating with other NIH Institutes and Centers and other federal agencies to support health activities such as the Family History Initiative and newborn screening. Additionally, the staff supports, participates in, and initiates informatics research projects at LHNCBC.

STATUS

The GHR web site became operational in April 2003 with about a dozen health conditions plus their related genes. After 2 years of operation, the content has steadily grown to include all human chromosomes, 7 chapters of the *Help Me Understand Genetics Handbook*, and more than 300 condition and gene summaries. As of March 2005, nearly 100 MedlinePlus topics were linked to GHR (see Table 2).

Web site traffic and anecdotal evidence indicate increasing usage and favorable acceptance of the GHR site. As shown in Figure 2, site traffic has increased fourfold over the past 2 years. Table 3 lists selected favorable comments made by healthcare providers, patients, family members, educators, librarians, and the press. Recently, online media outlets such as Forbes.com, CNN, and Nature.com have linked to GHR to provide background material for genetics-related stories.

In general, health conditions are viewed most often, about twice as frequently as the *Help Me Understand Genetics Handbook* and glossary, which are viewed next in frequency. Gene summaries, search results, general site pages such as the home page, chromosome summaries, and browse follow in usage (see Table 4). The small number of visits to chromosome summaries may be due to fewer GHR health topics linked to chromosome summaries than gene summaries. This hypothesis is supported by an increase in visits to chromosome summaries as more chromosomal conditions were added. Also, although more sessions start at a browse page rather than a search page, users are more likely to use the search function to find subsequent information.

Another visible trend is the larger number of visitors to the GHR web site during the week than on the weekends. Statistics on the types of web browsers accessing GHR each day suggest that the increase in traffic during the week is due to access from work or school as opposed to access from home.

METHODS AND PROCEDURES

GHR's methods and procedures are designed to create a comprehensive, reliable, easy-to-use resource to explain the effects of genetic variation on human health. Due to the large number of known genetic conditions, the first challenge is to prioritize topic selection. The second challenge is to streamline content development while ensuring accuracy. Third, GHR must help the lay public navigate the complex relationships between health conditions and genetic factors. Finally, because new data and knowledge relating the human genome to health continue to emerge, GHR must research new ways to meet ongoing and future challenges. Known and innovative informatics techniques developed at LHNCBC have been applied and will continue to support future GHR development.

Topic Selection Strategies

Approximately 1,700 heritable disorders (such as cystic fibrosis and sickle cell anemia) are known to be caused by mutations in single genes [15]. Most of these disorders are rare [4], and it can be difficult to find consumer-friendly information about them. Many other disorders (such as breast cancer and Parkinson disease) are multifactorial; they have a genetic component, but are also influenced by environmental factors and lifestyle choices [4]. These multifactorial conditions are often more common than single-gene disorders, but they are also more complex and their etiology is not completely delineated.

With so many heritable conditions from which to choose, GHR uses a multifaceted approach to prioritize topic development. This combination of strategies allows for both breadth and depth in the collection of topics that GHR ultimately presents to the public. First, GHR staff monitors genetics topics of national import, such as the recent Department of Health and Human Services newborn screening initiative [16]. As part of this initiative, a national committee of experts in genetics and public health developed a recommended panel of conditions to include in statewide newborn screening programs. The draft recommendations were released in March 2005. GHR plans to support the newborn screening initiative by developing summaries to cover all 29 genetic conditions identified by the expert committee. As of March 2005, GHR includes condition and gene summaries covering 20 of the 29 newborn screening topics (See Table 5). The remaining topics are in development or on GHR's high-priority topic development list.

GHR staff also coordinate with other federal government projects related to genetics, such as the Genetics and Rare Diseases Information Center [17], and prioritize content development to support these initiatives. In March 2005, staff at the Genetics and Rare Diseases Information Center, established by National Human Genome Research Institute (NHGRI) and the Office of Rare Diseases (ORD), provided a list of 20 disorders for which information is most often requested. GHR summaries are currently available for half of these disorders; all of the other conditions with a known genetic cause have been added to the GHR high-priority topic development list. By coordinating with the Genetics and Rare Diseases Information Center,

GHR can prioritize topic development in a way that is responsive to the public's information needs.

Another strategy for prioritizing the large number of potential genetics topics is to use MedlinePlus [12] as a guide. Many of the hundreds of MedlinePlus health topics are directly related to a heritable condition or have a genetic component. GHR aims to support all relevant MedlinePlus topics with related condition and gene summaries. Some broad health topics include several GHR topics; for example, GHR has developed summaries for more than 10 skeletal disorders to support the MedlinePlus health topic on "dwarfism." Other health topics are more specific and include a single GHR topic; for example, one GHR condition summary supports the MedlinePlus health topic on "Turner syndrome."

GHR staff maintain a list of all genetics-related MedlinePlus health topics to prioritize upcoming topics for the GHR web site. A status analysis of this list is performed annually because both GHR and MedlinePlus continually add new data. As of March 2005, GHR offers condition and gene summaries related to 92 MedlinePlus health topics (see Table 2). Each GHR topic links to related MedlinePlus health topics, and MedlinePlus topics automatically link back reciprocally to GHR. Additionally, MedlinePlus and GHR staff members collaborate regularly to develop complementary content for the two web sites. Using a comprehensive source of health information like MedlinePlus to prioritize upcoming topics allows GHR to develop a broad spectrum of genetic disorders.

GHR staff consider emerging disciplines in the intersection of genetics and health when planning future topic domains. These subject areas will require new paradigms of information management and presentation. Possible subject areas include mitochondrial DNA; the genetics of complex, multifactorial disorders; haplotype implications; epigenetics; pharmacogenomics; nutrigenomics; and environmental genomics. These areas are discussed in the Project Schedule section.

Streamlining Content Development

Several informatics strategies make GHR's content development process more efficient. All are based on the principle that automated data extraction is not perfect. Thus, tools can be created to assist in locating and collating relevant information, but all data must be reviewed by a qualified expert before they are presented to the public.

The foundation for the informatics used in GHR is collecting structured data instead of document-based information. GHR internally stores relational data and uses it to create a document-based presentation for the public. Elements in the data structure can be linked with other data sources. For example, the gene location code in GHR can be linked with the gene location code in Entrez Gene [2]. Linking data elements to other data sources provides the foundation for streamlining content development and maintenance. Structured data also supports the search feature to favor specific kinds of search result such as health conditions.

Many of GHR's strategies are based on mining information from existing research, clinical, and consumer databases such as Entrez Gene [2], HUGO Gene Nomenclature Committee (HGNC) [18], MedlinePlus [12], NCBI Map Viewer [19], Gene Ontology (GO) [20], GeneCards [21],

Gene Reviews and GeneTests [1], PubMed [22], and OMIM [15]. In addition to using the references inherent in some of these data, GHR uses semantic information from the Unified Medical Language System (UMLS) [23] to help join relevant information across multiple sources. For example, Entrez Gene and HGNC data can be easily joined using the Entrez Gene ID present in every HGNC record. Joining MedlinePlus to Gene Reviews, however, is not as easy. Semantic tools such as searching with expanded synonymy, indexing to NLM's Medical Subject Headings (MeSH), and extending a search using MeSH relationships help join records describing similar concepts.

The process used to create a gene record in GHR illustrates how data mining streamlines the content development process. To create a new gene record, the content developer enters a gene symbol into the GHR Content Manager (the software used to collect and store GHR's data). The software searches data from Entrez Gene [2] and HGNC [18] to find the gene's data. Often, many candidates are found. These possibilities are presented to the content developer to choose the correct match. If a match is selected, the software automatically prefills the gene symbol, name, location code, terms from GO [20], synonyms, and links to Entrez Gene [2], OMIM [15], GeneCards [21], and HGNC [18].

Data mining also streamlines the process of finding online resources related to particular conditions, genes, or chromosomes. GHR searches downloaded data sets and uses the NLM Gateway to suggest links to online resources such as MedlinePlus health topics and encyclopedia entries [12], OMIM topics [15], and Gene Reviews and GeneTests [1]. Mapping each GHR condition to a concept in MeSH improves the accuracy of the search algorithm. The MeSH mapping augments GHR's synonymy for condition names and acts as a bridge when other data sources also index to MeSH. In particular, MedlinePlus topics and PubMed [22] articles are both indexed to MeSH.

Another strategy is automatic translation between technical notation and a presentation understandable by the lay public. For example, researchers use a coded notation to denote the location of a gene. Without prior training, it is difficult to decipher the code. GHR's software automatically translates a gene location code, such as 12q13, into a sentence. For example, "The AAAS gene is located on the long (q) arm of chromosome 12 at position 13." Even with this translation, a lay-person may not understand what the long arm of a chromosome means. To help with this problem, GHR uses human genome map data available from the NCBI Map Viewer [19] to create an ideogram image of chromosome 12 that indicates the gene location.

Two strategies are used to facilitate GHR's glossary features. First, definitions are extracted from the UMLS [23] and from the GeneTests [1] illustrated glossary and presented in GHR's Content Manager. Content developers can search and choose from both of these data sets. Definitions from other sources such as the National Cancer Institute or MedlinePlus may also be added. Second, as part of creating the document-based presentation for the public site, GHR automatically searches each gene, condition, or chromosome summary for available glossary terms. The software automatically inserts links to all relevant glossary terms in each summary.

Ensuring Accurate and Current Information

The field of medical genetics encompasses a vast amount of information, which has grown larger as a result of the Human Genome Project [24]. These data are continually changing and being refined as researchers learn more about the complexities of the human genome. Therefore, to be useful, it is essential for any genetics web site to present information that is both accurate and current [9]. GHR uses a combination of expert review and automation to ensure that its condition and gene summaries are correct and up-to-date.

Each GHR condition and gene summary is fully reviewed for accuracy by an expert in the field of genetics before it is initially posted to the web site. GHR's reviewers typically have an M.D., Ph.D., or other advanced degree and are affiliated with a medical center, university, or laboratory. A list of GHR's expert reviewers and their affiliations is available at http://ghr.nlm.nih.gov/ghr/ExpertReviewers. Genetics experts also help GHR maintain up-to-date information. Each condition and gene summary undergoes an annual review by GHR staff and a genetics expert. The date of last comprehensive review is included at the bottom of each summary, so users will know that the information is current.

In addition to expert review, GHR uses several informatics strategies to ensure the presentation of up-to-date, reliable data. Automation increases GHR's ability to keep information synchronized with the latest available information from the Human Genome Project. Tools compare GHR data with data downloaded weekly from Entrez Gene [2], HGNC [18], MedlinePlus [12], and GO [20]. The tools identify differences that may indicate advances or changes in scientific understanding. These differences are assessed and corrections to GHR data are made as needed by staff.

One example is GHR's method for maintaining gene names and symbols. GHR relies on the official gene symbols and names designated by HGNC [18]. Over the past 2 years, HGNC has changed the official symbol and name of several genes described in GHR. Instead of relying on an annual expert review to identify and correct the names, GHR software checks gene symbols and names each week and notifies GHR staff of any changes. Thus errors can be corrected and posted to the public site within 2 weeks. Similar methods are used with other data sets to find new articles related to specific genetic conditions, genes or chromosomes; new or changed OMIM records [15]; new synonyms; new MedlinePlus topics; and new or changed Entrez Gene records [2]. Automation also assists with workflow-related tasks such as identifying condition, gene, and chromosome summaries that require an annual review.

Computerized methods also help maintain links from GHR web pages to other web sites. To prevent broken links, GHR software frequently tests the status of every external link in GHR and reports errors to GHR staff for remediation. Although the GHR system cannot discriminate between a site that has been removed from the Internet and one that is temporarily unavailable, it does find many problems and minimizes the manual work needed to maintain links.

Much of GHR's research focuses on methods to improve automation, which streamlines content development and maintenance. The GHR project team continually investigates new bioinformatics databases to enhance automation and extract relevant genetics information for developing content.

Helping Consumers Find Relevant Information

Web-based genetics information can be difficult to navigate because a consensus for naming and classifying genes and conditions is still emerging; a single disorder or gene may have different names in the scientific literature and among various resources. GHR helps the public retrieve information on topics of interest in genetic health and identify sources of related information. The search, browse, and linking features on the GHR web site allow users to locate information about a particular disorder or gene, using any of the naming possibilities. A unique feature of GHR is the clear presentation of associations between individual health conditions and related genes or chromosomes. This information spans a range of detail, from general to specific, to address consumer queries. GHR translates the health implications of Human Genome Project research into lay language, providing a comprehensive user-friendly and nontechnical resource for consumer-level genetic information.

A previous study [25] of consumer search behavior revealed that users primarily search NLM resources for information related to health conditions. Terminology-related issues, such as misspellings and abbreviations, are a primary obstacle to finding relevant information. GHR tunes search algorithms to give precedence to search results describing health conditions. Also, the site leverages infrastructure developed for ClinicalTrials.gov [13] to overcome terminology-related search issues [26]. Sharing this search infrastructure allows GHR to benefit from solutions to shared problems, such as inconsistent disease naming and classification, misspellings, and acronyms with multiple meanings. A reciprocal benefit to ClinicalTrials.gov is GHR's contribution of information about terminology specific to genetics.

GHR also includes search results from MedlinePlus [12], GeneTests [1], and Entrez Gene [2]. This is especially helpful when a user searches for a topic not yet available in GHR. For example, if a user searches for "juvenile diabetes," the search results offer a link to the MedlinePlus page about this condition. Similarly, when a user searches GHR for a gene that the site does not yet include, an annotated version of Entrez Gene data is presented. Because Entrez Gene data are challenging for the lay public, GHR truncates the presentation to include only information that is immediately relevant and understandable. For example, the presentation does not include sequence information, but does include gene product names, alternate symbols, and other aliases. If a user is interested in more detail, a link to the appropriate page in Entrez Gene is easily accessible. GHR also annotates the gene data with explanations and links to relevant explanations in the *Help Me Understand Genetics Handbook*. Presenting search results from other sites helps users navigate to other valuable resources when GHR content is not currently available.

GHR provides several types of browse features that allow users to find condition, gene, and chromosome summaries using alphabetical lists of names, gene symbols, or chromosome numbers. Alphabetical lists include the primary name chosen for GHR topics as well as synonyms. Hierarchical browse features are provided for condition and gene summaries. The browse hierarchy for a condition is loosely based on the relationships between MeSH condition concepts. For example, GHR's cancers category roughly maps to MeSH's neoplasms concept. The browse hierarchy for a gene is automatically derived from upper levels of GO [20]. For example, the GO hierarchy shows that both the APOE gene, which is associated with Alzheimer

disease, and the TPO gene, which causes one form of congenital hypothyroidism, have a molecular function related to antioxidant activity.

From a condition, gene, or chromosome summary, links are available to related information. Each summary includes links in the body of the text as well as a list of quick-navigation links along the left side of the page. Of particular interest is the relationship of genes or chromosomes to each condition. GHR uniquely delivers a consumer-focused explanation of the genes or chromosomes that are related to each health condition, how they are related to the condition, and which characteristics of the gene or chromosome affect the condition. GHR's presentation facilitates navigation among these factors to promote understanding of these important relationships. In addition, GHR links to explanations of relevant genetics concepts in the *Help Me Understand Genetics Handbook* and to other reliable web sites. Links to other web sites are selected for a wide range of audiences including patients, family members, clinicians, educators, students, and genetic researchers. When presenting these links, GHR groups them to help prepare the reader for the kind of information available from each source. At a glance, a user is able to see that navigating a link to OMIM will result in more challenging information than navigating a link to a patient support resource.

Research

GHR conducts applied research on how to present and develop complex medical information for lay use. GHR's content development process provides a working laboratory for research in the development of materials for consumer use. The LHNCBC is in an excellent position to conduct research in this area. Its active research programs in terminology issues, natural language processing, knowledge representation, information retrieval, health communication, and information systems provide relevant expertise and collaboration for the GHR project. For example, GHR uses NLM semantic tools [27] and vocabulary resources, including the UMLS [23], to help write and maintain content. GHR contributes genetic expertise to vocabulary resources. For example, GHR was a major motivation for incorporating GO into the UMLS [28].

As the GHR team moves forward, it will pursue opportunities to conduct formal research in synergistic areas with other LHNCBC groups. Important issues for GHR encompass many of the areas of research already being done at the Center. Information retrieval is one such area as GHR faces challenges in retrieving information to use in producing content, and retrieving information for intended users. Related to information retrieval is spelling suggestion or correction, which is especially important for finding genetics information with developing terminology and inconsistent naming conventions. GHR may contribute to vocabulary research by studying which lay terminology is best suited for presenting complex medical information to the nonexpert user. Vocabulary research in the burgeoning problem of gene names and synonyms in various species is also highly relevant to GHR. Projects in the natural language area, including summarization and question and answering research, are important for abstracting complex information into understandable and more manageable text. Semantic tools can be used for mining text from literature to help in choosing topics, and in developing and updating GHR content. Consumer health research, including readability and measurement of consumer understanding and satisfaction, are relevant areas of work for this project. Many opportunities exist for GHR to contribute to LHNCBC's research-oriented objectives.

EVALUATION

Before 2004, informal surveys of first-year college students and online users were conducted to help assess early layout and navigation designs, as well as content substance, level, and understandability. Feedback from these surveys helped shape glossary content, the question and answer layout of summaries, the placement and types of links, the prioritization of search results, and the *Help Me Understand Genetics Handbook*. System-level tests to ensure compliance with accessibility regulations and to stress system capacity were also conducted.

In early 2004, a survey was conducted to provide data for a more thorough consumer-based evaluation of the GHR web site. Members of the Genetic Alliance, an international coalition that represents individuals with genetic conditions, were selected as survey participants to provide a critical assessment of the site. Significant problem areas in navigation or content identified in the survey could be fixed before evaluating a larger, randomized population.

The objectives of the 2004 evaluation were to (1) obtain systematic consumer feedback about GHR, (2) provide a report that presented an overview of the findings, (3) contribute to consumer health informatics research literature, and (4) use the analyses to prepare a more comprehensive future evaluation. The online survey was conducted for seven weeks from late February to mid April, 2004. Participation was voluntary, and 374 respondents completed the survey.

The discussion of the 2004 evaluation here is abridged from three analyses that are included as supplementary material with this report [29, 30, 31].

Evaluation Methods

From its inception, GHR's 2004 evaluation was underpinned by a conceptual foundation developed last year by LHNCBC's consumer health informatics unit [32]. The conceptual foundation, which is reproduced in Figure 3 below, provides a more comprehensive, interactive, and multidisciplinary view of the process and effects surrounding the consumer health informatics research environment. As Napoli [33] and Friedman [34] noted, the array of research about consumer health informatics often has been more descriptive than analytical. Napoli [33] emphasized the need to build theory from the variety of disciplines (such as health communication and computer-mediated communication) to understand why consumers converge on a health informatics web site. In a recent paper that details LHNCBC's conceptual framework, Tse and Logan [35] agree that a key component of the evaluation of consumer health information web sites is to base research on consumer perceptions.

With this conceptual framework in mind, the GHR evaluation instrument focused on the user/consumer perceptions of GHR. A multidimensional approach was used, derived from the literature in consumer health informatics, mass communication, health communication and information science. The instrument was designed to enable consumers to provide feedback about GHR's content, design, and interface. As a result, the instrument was intended to yield user impressions for the GHR staff to consider in improving the site's usability and content. The instrument also supports additional research beyond a traditional descriptive analysis of a consumer health web site's demographic, usability, and utility of its central features. For example, the instrument advanced an exploration of the interaction between the complex

independent and dependent variables identified as relevant to understanding how and why users are motivated to use a consumer health web resource.

User perceptions of GHR were evaluated in four broad areas: (1) specific features; (2) usability, credibility, and users' post-use intentions and satisfaction; (3) demographics; and (4) GHR's image. (The survey instrument is included in Appendix A).

Evaluation Findings

The demographics of the survey participants were skewed towards female, well-educated, and experienced Internet users. About 78 percent of the persons surveyed had a college education or higher; 82 percent were female, and about 75 percent use the Internet more than 2 hours a day. In addition, most respondents who visited the site were family members or friends of a patient. The findings are not generalizable to all Genetics Alliance members or the general population because of the nonrandom nature of the survey.

The finding that the respondents predominately were female, well-educated, and sought healthcare information on the web in a nonprofessional capacity was similar to the profile of health Internet seekers that Pew recently identified [36]. This suggests the study's Genetic Alliance respondents may provide insights that are applicable to GHR users.

The survey results indicated that GHR's perceived credibility and overall consumer satisfaction were high [29]. Respondents also found GHR to be authoritative, accurate, unbiased, pertinent, up-to-date, and informative. The overall user satisfaction among the 374 respondents was very high; 88 percent of users surveyed said they were satisfied or very satisfied with GHR. This satisfaction suggests that the site appeals to a core audience, and that additional users may develop an interest in GHR.

Peng and Logan [30] explored independent variables that predict user satisfaction, as well as how users evaluated GHR's affective dimension. Their findings, which perceived content quality was a significant, strong predictor of both users' affective evaluation and overall satisfaction, are consistent with previous findings that consumers evaluate online health information predominantly based on their perception of several crucial elements of content quality in which credibility is key [36, 37, 38, 39].

Additionally, Peng and Logan [30] found no significant associations between prior online experience, prior interest and knowledge, and affective evaluations and overall consumer satisfaction. This observation appears to deviate somewhat from theoretical assumptions and findings in previous studies, which suggests consumer interest in health information is a strong positive predictor [40].

Additional analysis revealed three different prisms by which the survey respondents perceive GHR [31]. One prism reflects a perceptual orientation that views the overall design and visual appeal of GHR. A second prism is based on the site's perceived source credibility and quality of information. The third prism views GHR more by its perceived complexity/simplicity and the site's perceived bias in lieu of other reasons.

The findings of overall, high respondent satisfaction with GHR strongly suggest that even when persons agree, there are significant perceptual differences about a health web site's image and a consumer's interest in using the web site. The finding that persons favorably disposed toward GHR use it for different reasons may help GHR's staff better tailor decisions to meet user needs. For instance, the findings suggest that GHR's text needs to be authoritative, accurate, up to date and pertinent. The design also needs to be appealing, and attention needs to be given to the personality the site communicates to viewers. A more critical audience of users may view the content in terms of whether it is consistent with how they interpret genetics information, and whether the selection of topics for GHR aligns with their expectations.

In addition, the survey results demonstrate that profiling users of a health information web site by evaluating its image helps unveil the personality that a web site communicates to users. Although some aspects of GHR's perceived image may be unintended, different attitudes are projected onto the web site—even when respondents are favorably predisposed. Underlying these attitudes are judgments that form audience expectations, which influence how users may judge the web site.

Evaluation Summary

Overall, satisfaction with GHR was strong. Respondents provided consistently high ratings of GHR's features and credibility. One analysis revealed the importance of consumer-derived measures of aesthetics, content quality and usability, and how these affect user satisfaction of an innovative web site that attempts to improve consumer access to complex biomedical information [30]. A second analysis explored why persons are attracted to GHR and how users evaluate the web site's image and perceive its personality [31]. The evaluation of how audiences assess a healthcare web site's image may provide a tool to obtain fresh insights about GHR, or any other health information web site. The assessment of a health information web site's perceived image, coupled with more traditional audience demographic and behavioral measures, provides a range of tools that reveal comprehensive insights into consumer expectations and behavioral orientations.

PROJECT SCHEDULE

During the next year, the GHR project staff intend to continue meeting the goal of adding four new topics to the public site each month. Staff will also perform annual reviews of the growing list of existing topics and keep the information current. The *Help Me Understand Genetics Handbook* will be expanded and updated as needed to support other genetics information presented on the GHR web site.

GHR will enhance automation to make the content development process more efficient and help users find the genetic information they need. GHR staff plans to begin researching new informatics techniques and new ways to incorporate cutting-edge areas in genetic health (as described in the future plans for genetic health subject areas section below). Mitochondrial disorders will likely be among the first new genetic subject areas to be added because they will fit well into GHR's existing data structure and knowledge paradigm. Staff may begin incorporating other, more complex subject areas into GHR by adding new questions or chapters to the *Handbook.*

GHR staff will continue to collaborate with NIH Institutes and Centers and other federal agencies. These mutually beneficial relationships will help increase awareness of GHR and support federal health activities, such as the Family History Initiative and newborn screening.

GHR staff will also plan new evaluation studies. Several different audiences may be used to investigate the questions posed in the plans for future evaluations.

Future Plans for Informatics Research

As GHR continues to improve and expand, it will be in a position to explore more challenging research problems [3]. Continued development of the site will guide the use of informatics techniques and drive new research to assist in selecting new topics, developing content, ensuring accuracy and currency, and helping consumers find the information they need. Text mining techniques [41, 42] can be used to find new information from NIH fact sheets, PubMed, and the news media. Summarization and question-answering research [43] that is underway at LHNCBC will help to focus and condense relevant information for content development. In collaboration with ontology and terminology researchers at the Center [44, 45], the GHR team will address issues in information presentation and retrieval. Consumer health research [46, 47] will continue to guide the project on the needs of intended users and whether they can find the information of interest. The dynamic nature of the GHR project provides the Center with a real and growing system for research and development. In collaboration with other groups at LHNCBC, GHR is a strategic resource for investigating how informatics techniques can help in the development and retrieval of consumer-level health information.

As GHR evolves over the next several years, the project will continue to respond to the latest developments in the fields of informatics and human genetics. In the next 2 to 3 years, GHR staff will work with other project teams at LHNCBC to find ways to use techniques such as vocabulary research and natural language processing to enhance GHR. Additionally, staff will seek out further opportunities for GHR's structure and content to apply, support, and motivate research by other groups at the Center. As more definitive information becomes available about genetic subject areas, such as complex disorders and nutrigenomics, GHR staff will design new user-friendly presentation models to accurately include information about these areas.

Future Plans for Genetic Health Subject Areas

The tools and techniques developed in the course of GHR informatics research will aid in presentation of an expanding domain of knowledge at the intersection of genetics and the health implications of research from the Human Genome Project. Making this information accessible to the GHR audience will involve exploring new data sources, integrating new techniques, and finding presentation models to accommodate additional subject areas. Several of these potential subject areas are outlined below.

Mitochondrial DNA. GHR will expand the topic content to include disorders related to mutations in mitochondrial DNA. This type of DNA resides in mitochondria, the structures that produce energy for the cell. Mitochondrial DNA is distinct from the DNA in the cell nucleus. The circular arrangement of mitochondrial DNA contrasts to the linear structure of nuclear DNA, and the two types of DNA differ in how they are passed along during cell division. GHR's

current data structure and presentation should easily accommodate information in this subject area, although discussions of maternal inheritance patterns will need to be added.

Common, complex disorders. The complex nature of common disorders such as asthma, diabetes, autism, and various cancers creates content development and maintenance challenges for GHR. The contributions of many genes, environmental factors, and lifestyle choices can all affect an individual's risk of developing these disorders. Knowledge about the role genes play in these disorders is often not clear, and data are often conflicting. GHR has already included a few complex disorders, such as Parkinson disease, with a manageable number of related genes. These condition summaries acknowledge the partial role played by genetics and explain the specific genes thought to be associated with the disorder. GHR continues to explore new data sources and methods to meet these challenges.

Haplotype implications. As the International HapMap Project [48] nears completion, the health risks associated with specific haplotypes will become more apparent. A haplotype is the set of sequence variations along a particular region of a chromosome. Studies indicate that haplotypes will provide important clinical information, such as the acceptance rate of tissue and organ transplants [49], but the clinical implementation of this information is still emerging. GHR will consider how to collect, maintain, and present information on these new relationships as they solidify.

Epigenetics. Changes to DNA other than changes in the DNA sequence are called epigenetic factors. These changes affect how genes are expressed. For example, an epigenetic change may turn a gene on or off, or label the parental origin of a gene. These changes can be heritable and expressed in the sperm or egg, or nonheritable and expressed in somatic cells, often as a trigger for the growth of a malignant or benign tumor. Epigenetics is becoming increasingly important in understanding how nongenetic factors affect disease. The *Help Me Understand Genetics Handbook* currently discusses epigenetics in the context of genomic imprinting and uniparental disomy. Advances in epigenetics research will guide GHR's treatment of this subject.

Pharmacogenomics and drug/gene interactions. Recent news reports have highlighted genetic differences affecting response to drugs for common conditions such as hypertension and cancer, or to commonly used anesthetic drugs. The *Help Me Understand Genetics Handbook* currently explains the basic concept of pharmacogenomics with links to further information. This topic is getting considerable attention because of the pending release of test panels by Roche Pharmaceuticals and ParAllele BioScience targeted at genotyping patients for the common genetic variations associated with metabolism of many prescription and over-the-counter medications. As more definitive information becomes available, GHR will consider an expanded discussion of this subject.

Nutrition and genomics. Nutrigenomics [50] is the study of genetic differences in the way nutrients are metabolized and affect disease risk. It is of great interest to the public, as some individuals remain apparently healthy on a diet that predisposes others to heart disease, diabetes, and obesity. Many of the metabolic conditions targeted by the newborn screening programs involve dietary modifications and can be considered examples of nutrigenetics. Another example of the relationship between nutrition and genetic variations involves the MTHFR gene

and risk of neural tube defects such as spina bifida [51]. Women with particular variations in the MTHFR gene have an increased risk of delivering an infant with a neural tube defect. A number of studies, however, have documented that supplemental folic acid lowers the risk of neural tube defects among infants born to women with these gene variations [51]. As additional concrete examples of gene-nutrient interactions are discovered, they will guide GHR's development of this subject area.

Environmental genomics: Environmental genomics is the study of genetic differences and how various environmental agents affect disease risk. It remains unclear why some people develop disease when exposed to environmental agents while others remain healthy. Research in environmental genomics, however, has revealed that genetic variations can alter a person's ability to respond to environmental stress and their subsequent risk of disease. For example, innate immunity is thought to play a role in atherogenesis. Common mutations in the TLR4 gene are associated with differences in the inflammatory response to bacterial lipopolysaccharide [52]. A particular TLR4 polymorphism (Asp200Gly) is associated with a diminished inflammatory response to lipopolysaccharide and also appears to be associated with a decreased risk of atherosclerosis (but an increased susceptibility to severe bacterial infections) [53]. GHR will explore how to integrate findings in environmental genomics.

Future Plans for Outreach and Collaboration

After 2 years of operation, GHR staff have built a critical mass of content and are ready to focus on bringing GHR to the attention of potential users. Improved cross-linking between GHR and websites operated by other organizations within the National Institutes of Health may help interested users find GHR. Additionally, publishing and presenting to a broader spectrum of professional and lay groups will increase awareness of this resource. For example, a presentation to the American Academy of Pediatrics would help pediatricians learn that GHR has content relevant to families of infants who test positive for a heritable condition via newborn screening.

In addition to these well-defined strategies, GHR staff are discussing other possible outreach and collaboration ideas. For example, GHR staff could explore ways to release the structured data that underlies the GHR public site to genetics or informatics researchers. Another idea is to undertake an outreach program similar to the Information Rx project currently under way using MedlinePlus to help healthcare providers, patients, and caregivers utilize GHR more effectively.

Future Plans for Evaluation Analyses and Studies

Additional research issues can be explored from the 2004 GHR survey data set. Medical professionals, patients, and the general public may differ in their information-seeking behavior and evaluation of the web site. Further scrutiny into the differences may be helpful to tailor specific information for GHR’s target audiences.

Future evaluation of the GHR web site and its users may add new outcome variables. Also, some of the variables used in analyzing the data from the 2004 GHR survey may be augmented to create constructs based on a cluster of more than seven questions. (See Table 6 for lists of potential outcome variables.) All of these outcome variables, identified in an ongoing review of the literature at LHNCBC [54], were used in recent consumer health informatics research.

Future surveys among motivated health seeking Internet consumers and persons motivated to seek genetics information via the Internet will clarify GHR's perceived readability, usability and image. Evaluations among special populations of potential GHR users may shape and guide GHR's support for targeted initiatives. For example, a special population of healthcare providers or affected families may help GHR support the existing government initiative on newborn screening.

SUMMARY

GHR is a credible, comprehensive, and dynamic web site that uses lay language to explain the effects of genetic variation on human health. The GHR project supports many of the goals of NLM's long range plan, particularly by providing "access to health information that is useful both to the general public and to practitioners who need information outside their particular field of expertise." LHNCBC offers an ideal setting to address consumer education in genetics. The innovative techniques developed through informatics research at LHNCBC are key to the effective management of a large and expanding body of genetics information.

Feedback from formal and informal evaluations have helped shape GHR's layout, navigational design, and level of content. Continued development of the site will guide the use of informatics techniques and drive new research to assist in selecting new topics, developing content, ensuring accuracy and currency, and helping consumers navigate the complex world of genetics.

ACKNOWLEDGEMENTS

Alexa T. McCray, former Director of LHNCBC, was instrumental in moving GHR into production and fostering its development during the crucial incubation period. She also assisted tremendously in the human factors and navigational design of the site. Her leadership during the early phases of GHR laid the foundation for a high-quality, useful consumer health resource.

We would like to acknowledge the contributions of Nicholas Ide, who provides insightful advice on system architecture, the tools available at LNHCBC, search tuning, the web server, and operational configuration; John Gillen, who implements and maintains the GHR Content Manager; Russell Loane, who implemented the data layer in the Content Manager and developed the search engine; Erik Dorfman, who provided configuration management and system components for managing controlled vocabularies (such as MeSH); and Kelly Zeng, who implemented the glossary content management feature.

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REFERENCES

1. University of Washington, Seattle. GeneTests. [Online] Available http://www.genetests.org/. Last accessed March 2005.

2. U.S. National Institutes of Health, National Library of Medicine, National Center for Biotechnology Information. Entrez Gene. [Online] Available http://www.ncbi.nih.gov/entrez/query.fcgi?db=gene/. Last accessed March 2005.

3. Mitchell JA, McCray A, Bodenreider O. From phenotype to genotype: issues in navigating the available information resources. Methods Inf Med 2003; 42(5):557-63.

4. Guttmacher, Alan E; Collins, Francis S; Drazen, Jeffrey M. Genomic medicine : articles from the New England journal of medicine. Baltimore : Johns Hopkins University Press ; Boston : The New England Journal of Medicine, c2004. 3-14.

5. Guttmacher, Alan E; Collins, Francis S; Drazen, Jeffrey M. Genomic medicine : articles from the New England journal of medicine. Baltimore : Johns Hopkins University Press ; Boston : The New England Journal of Medicine, c2004. 166-171.

6. Khoury MJ. Genetics and genomics in practice: the continuum from genetic disease to genetic information in health and disease. Genet Med 2003; 5(4):261-8.

7. Cline RJ, Haynes KM. Consumer health information seeking on the Internet: the state of the art. Health Educ Res 2001; 16(6):671-92.

8. Fox S, Fallows D. Internet Health Resources. 2003. Washington, D.C., Pew Foundation. Pew Internet and American Life Project.

9. Guttmacher AE. Human genetics on the Web. Annu Rev Genomics Hum Genet 2001; 2:213-33.

10. Taylor MR, Alman A, Manchester DK. Use of the Internet by patients and their families to obtain genetics-related information. Mayo Clin Proc 2001; 76(8): 772-6.

11. Bernhardt JM, Lariesy RA, Parrott RL, Silk KJ, Felter EM. Perceived barriers to Internet-based health communication on human genetics. J Health Commun 2002; 7(4):325-40.

12. U.S. National Institutes of Health, National Library of Medicine. MedlinePlus. [Online] Available http://www.medlineplus.gov/. Last accessed March 2005.

13. U.S. National Institutes of Health, National Library of Medicine. ClinicalTrials.gov. [Online] Available http://clinicaltrials.gov. Last accessed March 2005.

14. U.S. National Institutes of Health, National Library of Medicine. Long Range Plan 2000-2005. [Online] Available http://www.nlm.nih.gov/pubs/plan/lrp00/toc.html. Last accessed March 2005.

15. U.S. National Institutes of Health, National Library of Medicine, National Center for Biotechnology Information and Johns Hopkins University. Online Mendelian Inheritance in Man (OMIM). [Online] Available http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM. Last accessed March 2005.

16. U.S. Department of Health and Human Services, Health Resources and Services Administration, Maternal and Child Health Bureau. Newborn Screening: Toward a Uniform Screening Panel and System. Executive Summary. [Online] Available ftp://ftp.hrsa.gov/mchb/genetics/screeningdraftsummary.pdf. Last accessed March 2005.

17. U.S. National Institutes of Health, National Human Genome Research Institute and Office of Rare Diseases. Genetic and Rare Diseases Information Center. [Online] Available http://rarediseases.info.nih.gov/html/resources/info_cntr.html. Last accessed March 2005.

18. HUGO Gene Nomenclature Committee (HGNC). [Online] Available http://www.gene.ucl.ac.uk/nomenclature/. Last accessed March 2005.

19. U.S. National Institutes of Health, National Library of Medicine, National Center for Biotechnology Information. Map Viewer. [Online] Available http://www.ncbi.nlm.nih.gov/mapview/maps.cgi?org=human. Last accessed March 2005.

20. Gene Ontology Consortium. Gene Ontology. [Online] Available http://www.geneontology.org/. Last accessed March 2005.

21. Weizmann Institute of Science. GeneCards. [Online] Available http://genecards.weizmann.ac.il/. Last accessed March 2005.

22. U.S. National Institutes of Health, National Library of Medicine. PubMed. [Online] Available http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=PubMed. Last accessed March 2005.

23. Humphreys BL, Lindberg DA, Schoolman HM, Barnett GO. The Unified Medical Language System: an informatics research collaboration. J Am Med Inform Assoc 1998; 5(1):1-11.

24. Collins DS, Green ED, Guttmacher AE, Guyer MS, US National Human Genome Research Institute. A vision for the future of genomics research. Nature 2003; 422(6934):835-47.

25. McCray AT, Loane RF, Browne AC, Bangalore AK. Terminology issues in user access to Web-based medical information. Proc AMIA Symp. 1999;:107-11.

26. McCray AT, Dorfman E, Ripple A, Ide NC, Jha M, Katz DG, Loane RF, Tse T. Usability issues in developing a Web-based consumer health site. Proc AMIA Symp. 2000;556-60.

27. Browne AC, Divita G, Aronson AR, McCray AT. UMLS language and vocabulary tools. Proc. AMIA Symp, November 2003;798.

28. Lomax J, McCray AT. Mapping the Gene Ontology into the Unified Medical Language System. Comp Funct Genom 2004:5:354-361.

29. Logan RA. Preliminary Report - Evaluation of Genetics Home Reference by Genetic Alliance. December, 2005.

30. Peng Z, Logan RA. Content Quality, Usability, Affective Evaluation, and Overall Satisfaction of Online Health Information, Accepted for presentation to Health Communication Division, International Communication Association Annual Meeting, May 2005.

31. Logan RA, Fun J, Cheh M. User Perceptions of Genetics Home Reference: A Study of a Health Information Web Site's Image as a Media Product. Submitted to American Medical Informatics Association 2005 meeting.

32. Tse T. Consumer Health Information Seeking: Report to the Board of Scientific Counselors, Lister Hill National Center for Biomedical Communications, May 13, 2004.

33. Napoli PM. Consumer use of medical information from electronic to paper media: A literature review. In: Rice Ronald E, Katz James E (eds). The internet and health communication: experiences and expectations. Thousand Oaks, Calif: Sage Publications, c2001.79-98.

34. Friedman CP. Toward a measured approach to medical Informatics. J Med Inform Assoc 1999; 6(2):176-7.

35. Tse T, Logan RA. Towards a More Comprehensive Conceptual Framework for Consumer Health Information Seeking. Submitted to American Medical Informatics Association 2005 meeting.

36. Pew Research Center. Internet health resources. Washington: Pew Internet and American Life Project 2003; p. 20-25. [Online] Available http://www.pewinternet.org/. Last accessed October 2004.

37. Fox S, Rainie L, Pew Research Center. The online health care revolution: How the Web helps Americans take better care of themselves. [Online] Available http://www.pewinternet.org/.

38. Baker L, Wagner TH, Singer S, Bundorf MK. Use of the Internet and E-mail for health care information: results from a national survey. JAMA 2003 May 14; 289(18):2400-6.

39. Eysenbach G, Powell J, Kuss O, Sa ER. Empirical studies assessing the quality of health information for consumers on the world wide web: a systematic review. JAMA 2002; 287(20):2691-700.

40. van der Rijt GAJ. Determinants of the consumption of health information in the media. Eur J Commun Res 1998;23(3):255-69.

41. Aronson AR. Effective mapping of biomedical text to the UMLS Metathesaurus: the MetaMap program. Proc AMIA Symp. 2001;17-21.

42. Rindflesch TC, Tanabe L, Weinstein JN, Hunter L. EDGAR: extraction of drugs, genes and relations from the biomedical literature. Pac Symp Biocomput. 2000;517-28.

43. Fiszman M, Rindflesch TC, Kilicoglu H. Abstraction summarization for managing the biomedical research literature. Proc HLT-NAACL Workshop Comp. Lex Sem. 2004;76-83.

44. Bodenreider O, Mitchell JA, McCray AT. Biomedical ontologies. Pac Symp on Biocomput. 2005;76-8.

45. Sarkar IN, Cantor MN, Bodenreider O, Lussier YA. GeneTrace(TM): Biological Knowledge Discovery via Structured Terminology. MedInfo.2004;20041842.

46. Logan RA. Evaluating consumer informatics: learning from health campaign research. Medinfo.2004;2004:147-51.

47. Soergel D, Tse T, Slaughter L. Helping healthcare consumers understand: an "interpretive layer" for finding and making sense of medical information. Medinfo 2004;2004:931-5.

48. International HapMap Project. [Online] Available http://www.hapmap.org/. Last accessed March 2005.

49. Crawford DC, Nickerson DA. Definition and clinical importance of haplotypes. Annu Rev Med. 2005;56:303-20.

50. Simopoulos AP, Ordovas JM (eds). Nutrigenetics and Nutrigenomics. Basel, Switzerland: S. Karger AG: World Review of Nutrition and Dietetics, vol 93, 2004.

51. Gibson KM, Bottiglieri T. Genetic Predisposition to Neural Tube Defects? Pediatr Res. 2000; 48(2):135.

52. Arbour NC, Lorenz E, Schutte BC, Zabner J, Kline JN, Jones M, Frees K, Watt JL, Schwartz DA. TLR4 mutations are associated with endotoxin hyporesponsiveness in humans. Nat Genet. 2000 Jun;25(2):187-91.

53. Kiechl S, Lorenz E, Reindl M, Wiedermann CJ, Oberhollenzer F, Bonora E, Willeit J, Schwartz DA. Toll-like receptor 4 polymorphisms and atherogenesis. N Engl J Med. 2002 Jul 18;347(3):185-92.

54. Bakker T, Ryce A, Logan RA, Tse T, Hutcherson L: A Consumer Health Informatics (CHI) Toolbox: Challenges and Implications. Submitted to American Medical Informatics Association 2005 meeting.

FIGURES

Figure 1: GHR Content Development Statistics

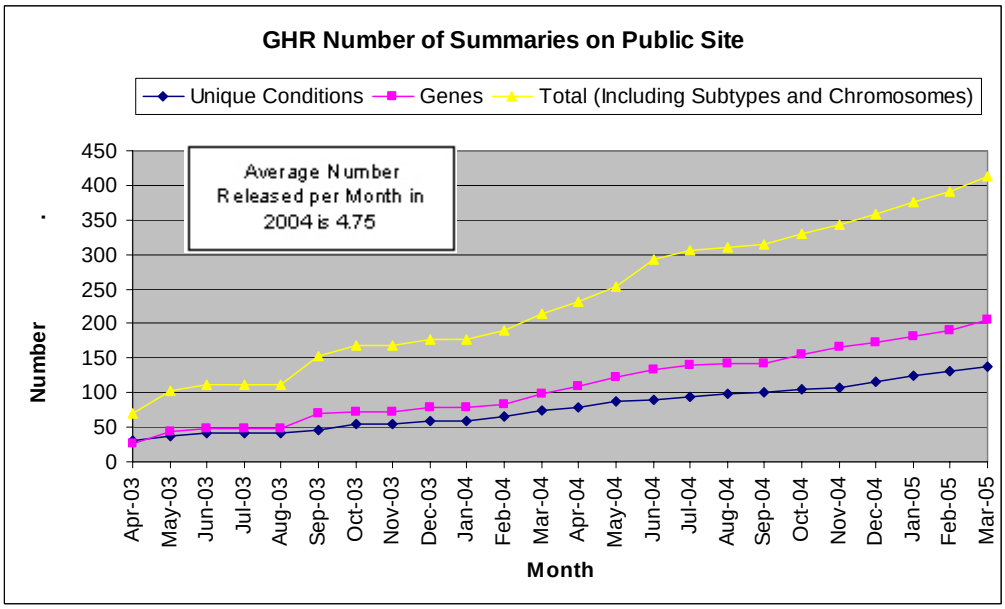


Figure 2: GHR Public Site Access Trends May 2003 to March 2005

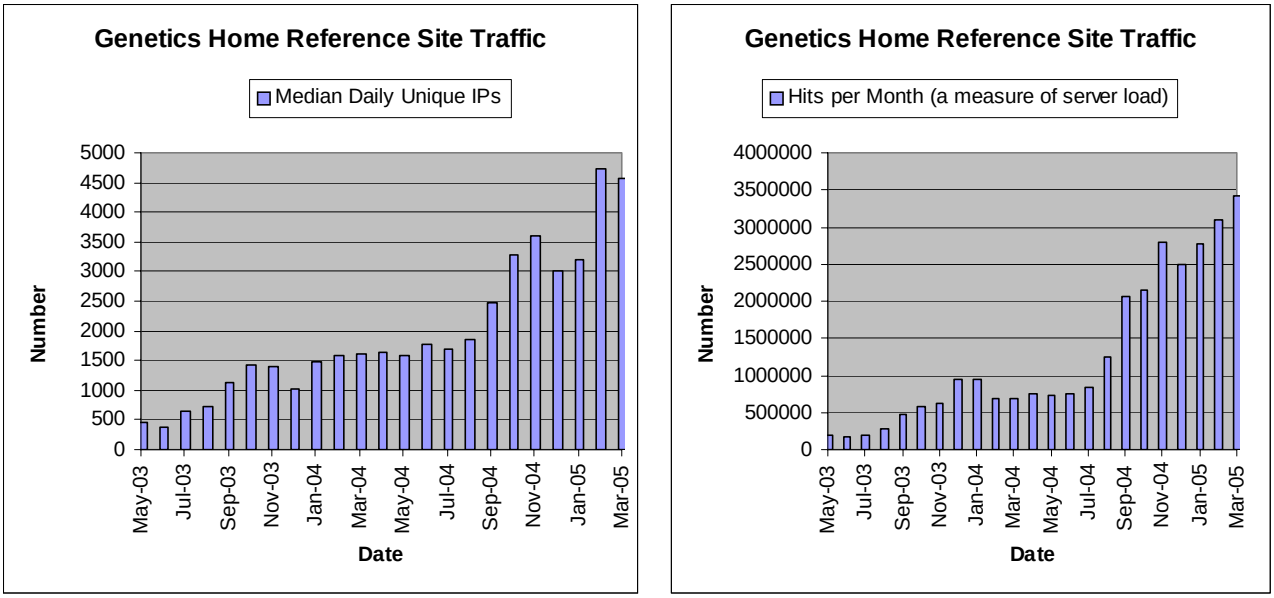
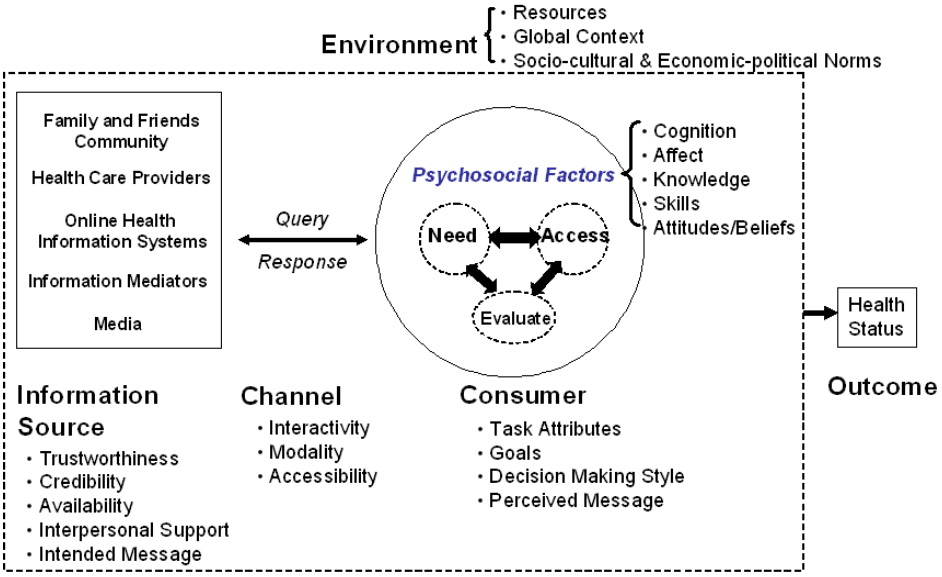


Figure 3: Consumer Health Information Conceptual Framework



Tse T, Logan RA. Towards a More Comprehensive Conceptual Framework for Consumer Health Information Seeking. Submitted to American Medical Informatics Association 2005 meeting.

TABLES

Table 1: GHR Condition, Gene, and Chromosome Summaries (March 31, 2005)

Conditions (138 + 51 Subtypes), Genes (228), Chromosomes (23 pairs)

GHR Conditions and Subtypes	GHR Genes, Chromosomes
Achondrogenesis, type 1B	SLC26A2
Achondrogenesis, type 2	COL2A1
Achondroplasia	FGFR3
Alport syndrome	COL4A3, COL4A4, COL4A5
Alagille syndrome	JAG1
Alexander disease	GFAP
Alkaptonuria	HGD
Alpha-1 antitrypsin deficiency	SERPINA1
Alström syndrome	ALMS1
Alzheimer disease (plus four subtypes)	APP, APOE, PSEN1, PSEN2
Amyotrophic lateral sclerosis (plus three subtypes)	ALS2, ALS4, NEFH, SOD1
Androgen insensitivity syndrome	AR
Andersen-Tawil syndrome	KCNJ2
Angelman syndrome	OCA2, UBE3A, Chromosome 15
Apert syndrome	FGFR2
Argininosuccinic aciduria	ASL
Ataxia-telangiectasia	ATM
Atelosteogenesis, type 2	SLC26A2
Beare-Stevenson cutis gyrata syndrome	FGFR2
Beta thalassemia	HBB
Biotinidase deficiency	BTD
Birt-Hogg-Dubé syndrome	FLCN
Bladder cancer	FGFR3, HRAS, RB1, TP53
Breast cancer	AR, ATM, BRCA1, BRCA2, CHEK2, DIRAS3, ERBB2, RAD51
CADASIL	NOTCH3
Canavan disease	ASPA
Charcot-Marie-Tooth disease (plus four subtypes)	EGR2, GARS, GDAP1, GJB1, HSPB1, KIF1B, LITAF, LMNA, MFN2, MPZ, MTMR2, NDRG1, NEFL, PMP22, PRX, RAB7, SBF2
Cockayne syndrome	ERCC6, ERCC8
Coffin-Lowry syndrome	RPS6KA3
Collagenopathy, types II and XI	COL11A1, COL11A2, COL2A1
Congenital bilateral absence of vas deferens	CFTR
Congenital hypothyroidism	PAX8, SLC5A5, TG, TPO, TSHB, TSHR
Cornelia de Lange syndrome	NIPBL
Cowden syndrome	PTEN

GHR Conditions and Subtypes	GHR Genes, Chromosomes
Cri-du-chat syndrome	Chromosome 5
Crouzon syndrome	FGFR2
Crouzonodermoskeletal syndrome	FGFR3
Cystic fibrosis	CFTR
Diastrophic dysplasia	SLC26A2
Distal spinal muscular atrophy, type V	GARS
Down syndrome	Chromosome 21
Edwards syndrome	Chromosome 18
Ehlers-Danlos syndrome (plus six subtypes)	ADAMTS2, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, PLOD1, TNXB
Fabry disease	GLA
Factor V Leiden thrombophilia	F5
Familial adenomatous polyposis	APC, MUTYH
Familial dysautonomia	IKBKAP
Familial lipoprotein lipase deficiency	LPL
Familial Mediterranean fever	MEFV, SAA1
Fragile X syndrome	FMR1
Friedreich ataxia	FXN
Galactosemia	GALE, GALK1, GALT
Gaucher disease (plus four subtypes)	GBA
Hemochromatosis (plus four subtypes)	HAMP, HFE, HFE2, SLC40A1, TFR2
Hemophilia	F8, F9
Hereditary neuropathy with liability to pressure palsies	PMP22
Hereditary nonpolyposis colorectal cancer	MLH1, MSH2, MSH6, PMS2
Homocystinuria	CBS, MTHFR, MTR, MTRR
Huntington disease	HD
Hutchinson-Gilford progeria syndrome	LMNA
Hyperphenylalaninemia	GCH1, PAH, PCBD1, PTS, QDPR
Hypochondrogenesis	COL2A1
Hypochondroplasia	FGFR3
Incontinentia pigmenti	IKBKG
Infantile-onset ascending hereditary spastic paralysis	ALS2
Jackson-Weiss syndrome	FGFR2
Jervell and Lange-Nielsen syndrome	KCNE1, KCNQ1
Juvenile primary lateral sclerosis	ALS2
Klinefelter syndrome	Chromosome X, Chromosome Y
Kniest dysplasia	COL2A1
Krabbe disease	GALC
Lesch-Nyhan syndrome	HPRT1
Li-Fraumeni syndrome	CHEK2, TP53
Long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency	HADHA

GHR Conditions and Subtypes	GHR Genes, Chromosomes
Maple syrup urine disease	BCKDHA, BCKDHB, DBT, DLD
Marfan syndrome	FBN1
Medium-chain acyl-coenzyme A dehydrogenase deficiency	ACADM
Menkes syndrome	ATP7A
Methemoglobinemia, beta-globin type	HBB
Methylmalonic acidemia	MMAA, MMAB, MUT
Mitochondrial trifunctional protein deficiency	HADHA, HADHB
Muenke syndrome	FGFR3
Multiple endocrine neoplasia type 1	MEN1
Multiple endocrine neoplasia type 2	RET
Muscular dystrophy, Duchenne and Becker types	DMD
Myotonic dystrophy (plus two subtypes)	DMPK, ZNF9
Neurofibromatosis (plus two subtypes)	NF1, NF2
Niemann-Pick disease	NPC1, NPC2, SMPD1
Nonsyndromic deafness (plus three subtypes)	CDH23, CLDN14, COCH, COL11A2, EYA4, GJB2, GJB3, GJB6, KCNQ4, MYO15A, MYO1A, MYO6, MYO7A, OTOF, PCDH15, POU3F4, SLC26A4, STRC, TECTA, TMC1, TMIE, TMPRSS3, USH1C, WFS1
Noonan syndrome	PTPN11
Osteogenesis imperfecta (plus four subtypes)	COL1A1, COL1A2
Otospondylomegaepiphyseal dysplasia	COL11A2
Pantothenate kinase-associated neurodegeneration	PANK2
Parkinson disease	LRRK2, NR4A2, PARK2, PARK7, PINK1, SNCA, SNCAIP, UCHL1
Patau syndrome	Chromosome 13
Pendred syndrome	SLC26A4
Peutz-Jeghers syndrome	STK11
Pfeiffer syndrome	FGFR1, FGFR2
Phenylketonuria	PAH
Prader-Willi syndrome	OCA2, Chromosome 15
Primary hyperoxaluria	AGXT, GRHPR
Primary pulmonary hypertension	BMPR2
Polycystic kidney disease	PKD1, PKD2, PKHD1
Porphyria (plus eight subtypes)	ALAD, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS
Prion disease	PRNP
Propionic acidemia	PCCA, PCCB
Pseudoxanthoma elasticum	ABCC6
Recessive multiple epiphyseal dysplasia	SLC26A2
Retinoblastoma	RB1

GHR Conditions and Subtypes	GHR Genes, Chromosomes
Rett syndrome	MECP2
Romano-Ward syndrome	ANK2, KCNE1, KCNE2, KCNH2, KCNQ1, SCN5A
Rubinstein-Taybi syndrome	CREBBP
SADDAN	FGFR3
Sickle cell anemia	HBB
Spinal and bulbar muscular atrophy	AR
Spinal muscular atrophy	SMN1, SMN2
Spondyloepimetaphyseal dysplasia, Strudwick type	COL2A1
Spondyloepiphyseal dysplasia congenita	COL2A1
Spondyloperipheral dysplasia	COL2A1
Stickler syndrome (plus two subtypes)	COL11A1, COL11A2, COL2A1
Tay-Sachs disease	HEXA
Tetrahydrobiopterin deficiency	GCH1, PCBD1, PTS, QDPR
Thanatophoric dysplasia (plus two subtypes)	FGFR3
Trimethylaminuria	FMO3
Triple X syndrome	Chromosome X
Tuberous sclerosis	TSC1, TSC2
Turner syndrome	SHOX, Chromosome X
Usher syndrome (plus three subtypes)	CDH23, MASS1, MYO7A, PCDH15, USH1C, USH1G, USH2A, USH3A
Very long-chain acyl-coenzyme A dehydrogenase deficiency	ACADVL
Von Hippel-Lindau syndrome	VHL
Waardenburg syndrome	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10
Weissenbacher-Zweymüller syndrome	COL11A2
Wilson disease	ATP7B
X-linked severe combined immunodeficiency	IL2RG
X-linked sideroblastic anemia	ALAS2, HFE
21-hydroxylase deficiency	CYP21A2
22q11.2 deletion syndrome	TBX1, Chromosome 22
47,XYY syndrome	Chromosome Y
--	Chromosome 1-4, 6-12, 14, 16, 17, 19, 20

Table 2: 91 MedlinePlus Topics Linked to GHR (March 31, 2005)

Adrenal Gland Disorders	Ehlers-Danlos Syndrome	Movement Disorders
Alpha-1 Antitrypsin Deficiency	Endocrine Diseases	Muscular Dystrophy
Alzheimer's Caregivers	Eye Cancer	Neurofibromatosis
Alzheimer's Disease	Facial Injuries and Disorders	Neurologic Diseases
Amyotrophic Lateral Sclerosis	Fever	Neuromuscular Disorders
Anemia	Fragile X Syndrome	Newborn Screening
Arrhythmia	Gaucher's Disease	Osteogenesis Imperfecta
Ataxia Telangiectasia	Genes and Gene Therapy	Parathyroid Disorders
Bile Duct Diseases	Genetic Brain Disorders	Parkinson's Disease
Birth Defects	Genetic Counseling	Peripheral Nerve Disorders
Bladder Cancer	Genetic Disorders	Phenylketonuria
Bleeding Disorders	Genetic Testing	Pheochromocytoma
Bone Diseases	Head and Brain Malformations	Porphyria
Bone Marrow Diseases	Hearing Disorders and Deafness	Prader-Willi Syndrome
Brain Diseases	Hearing Problems in Children	Pulmonary Hypertension
Breast Cancer	Hemochromatosis	Retinal Disorders
Cancer	Hemophilia	Sickle Cell Anemia
Cardiomyopathy	Huntington's Disease	Skin Diseases
Carpal Tunnel Syndrome	Immune System and Disorders	Skin Pigmentation Disorders
Charcot-Marie-Tooth Disease	Infertility	Speech and Communication Disorders
Cleft Lip and Palate	Kidney Cancer	Spinal Muscular Atrophy
Colorectal Cancer	Kidney Diseases	Stroke
Congenital Heart Disease	Klinefelter's Syndrome	Tay-Sachs Disease
Connective Tissue Disorders	Learning Disorders	Thyroid Cancer
Creutzfeldt-Jakob Disease	Leukodystrophies	Thyroid Diseases
Cystic Fibrosis	Lewy Body Disease	Tuberous Sclerosis
Degenerative Nerve Diseases	Liver Diseases	Turner's Syndrome
Dementia	Male Breast Cancer	Vision Impairment and Blindness
Developmental Disabilities	Male Genital Disorders	Wilson's Disease
Down Syndrome	Marfan Syndrome	
Dwarfism	Metabolic Disorders	

Table 3: Selected Praise from GHR Users

Date	Source	Comment
2/3/05	Course Page for BIOL 1103, Southern Wesleyan University	"The **best overall reference on human heredity** for students, and other non-geneticists, is the Genetics Home Reference, from the US National Library of Medicine."
12/27/04	E-mail: physician	"I found your genetics home reference to be **just what I have been searching for** for the past several years. **I will use your product as my genetics hub page**."
11/12/04	*Science Magazine*, Netwatch column	"This primer on genetic diseases from the U.S. National Library of Medicine can serve as a reference for students and help teachers catch up on the latest findings."
4/6/04	E-mail: family member of patient	"I have a son recently diagnosed with Noonan Syndrome. I found the website to be extremely helpful in explaining the genetics of the syndrome. **Despite extensive investigation on my part, this was the first time I have read information that was "parent friendly", yet detailed**. I learned much that I had not yet learned."
2/5/04	E-mail: unknown	"I wanted to let you know what a wonderful resource this is for public libraries! I just discovered it today and am delighted."
October 2003	Health Sciences Library System, University of Pittsburgh Medical Center	"Regardless of your level of understanding, **Genetics Home Reference has something for everyone**."
9/10/03	*Columbus Federal Voice*	"The site's **explanations are straight and simple, and written in easily understandable, jargon-free English**."
Summer 2003	NCI-Frederick, *Science Library News*	"Created in response to the increasing trend for the public to turn to the Web for medical information, the target audience is the general public and the language is written in simple and understandable English."
7/8/03	*The Washington Post*, health web site review	"The field of genetics is nearly as full of empty hype, commercial self-interest and hysterical predictions of human transformation as the Internet was in 1998. This **level-headed, science-based accounting of current knowledge from a credible source** is a useful counterweight."
[ongoing]	Aussie Educator, Tertiary Links, Genetics	"**A brilliant site for students as well as the general public**. The glossary, Genes & Conditions, 'Help me Understand ...' and the various resources are quite amazing, especially the links provided in the Resources' section which include a variety of databases."

Table 4: Visits to GHR Pages

Page Group	Visits to GHR in March 2005		
	Total	**First page in session[1]**	**Subsequent page in session[2]**
Condition*	125,642	59,355	66,287
Handbook**	55,303	12,544	42,759
Glossary	53,351	22,955	30,396
Gene	28,413	8,994	19,419
Search results***	25,540	1,273	24,267
General site****	25,155	11,576	13,579
Chromosome	14,519	4,888	9,631
Browse	11,707	1,992	9,715

* Conditions include group pages for linking to MedlinePlus, such as Dwarfism.
** *Help Me Understand Genetics Handbook.*
*** Search results include annotated Entrez Gene pages.
**** General site includes pages such as Home, Resources, Help, and What's New.
[1] The first page in a session is the first page a user views when accessing GHR
[2] The subsequent pages in a session are the pages a users views after the first page.

Table 5: Draft Recommended Core Newborn Screening Panel (March 2005)

Terminology from draft report	GHR condition name (Available conditions shown in **Bold-Green**)
Organic acid disorders (9)	
IVA	isovaleric acidemia
GA I	glutaric academia (type 1)
HMG	3-hydroxy-3-methylglutaryl coenzyme A lyase deficiency
MCD	multiple carboxylase deficiency
MUT	**methylmalonic acidemia**
Cbl A, B	**methylmalonic acidemia**
3MCC	3-methylcrotonyl coenzyme A deficiency
PROP	**propionic acidemia**
BKT	beta-ketothiolase deficiency
Fatty acid oxidation disorders (5)	
MCAD	**medium-chain acyl coenzyme A dehydrogenase deficiency**
VLCAD	**very long-chain acyl coenzyme A dehydrogenase deficiency**
LCHAD	**long-chain 3-hydroxyacyl coenzyme A dehydrogenase deficiency**
TFP	**mitochondrial trifunctional protein deficiency**
CUD	carnitine update disorder
Amino acid disorders (6)	
PKU	**phenylketonuria**
MSUD	**maple syrup urine disease**
HCY	**homocystinuria**
CIT	citrullinemia
ASA	**argininosuccinic acidemia**
TYR I	tyrosinemia
Hemoglobinopathies (3)	
Hb SS	**sickle cell anemia**
Hb S/ßTh	discussed in **HBB gene summary**
Hb S/C	discussed in **HBB gene summary**
Other (6)	
CH	**congenital hypothyroidism**
BIOT	**biotinidase deficiency**
CAH	**21-hydroxylase deficiency**
GALT	**galactosemia**
HEAR (a hearing test for hearing loss)	**nonsyndromic deafness, syndromic deafness disorders**
CF	**cystic fibrosis**

Newborn Screening: Toward a Uniform Screening Panel and System, Maternal and Child Health Bureau, Health Resources and Services Administration [http://mchb.hrsa.gov/screening/]

Table 6: Potential Outcome Variables for Future Evaluation Studies

New Outcome Variables	Variables Used in Evaluating GHR that could be Augmented
- Self-efficacy - Perceived empowerment - Health status - Patient-physician communication status - Perception of information provided by medical professional - Perception of receiving health information from the Internet versus print and broadcast media - Attitudes about health - Attitudes about health information seeking - Attitudes about health information seeking on the Internet - Cognitive load (of the web site) - Attitudes about religious faith versus genetic inheritance - Desire for control (about health and life) - Fatalism (about health and life) - Prevention orientation - Health services utilization - Perceived health services accessibility - Self-reported understanding of genetics - Behavioral outcomes - Health outcomes - Awareness of healthcare information availability	- Source credibility (cognitive dimension) - Affective dimension - Usability dimension - Health information seeking behaviors - Perceived readability of the health information web site - Perceived usability of the health information web site - Perceived use of features of the health information web site - Communication outcomes (improved communication with healthcare providers, other care givers, family members) - User satisfaction with a health information web site

These lists are not an exhaustive, but indicate the range of topics that could be explored and enhanced by future, comprehensive evaluations of health information web sites and their users.

APPENDIX A: GHR SURVEY INSTRUMENT

This is a reformatted version of the survey.

Survey Instructions

Please help us improve the health information that the Lister Hill National Center for Biomedical Communications at the National Library of Medicine (NLM) brings to you on the Genetics Home Reference web site by taking the following Consumer Satisfaction Survey. Completion of this survey is strictly voluntary and in no way affects any of your rights or privileges. We estimate that it should take no more than 25 minutes to read the instructions and complete the survey. NLM is required to inform you that no Federal agency may conduct or sponsor, and no member of the public is required to respond to, a collection of information unless it displays a currently valid OMB control number. For this survey the OMB Control Number is 0925-0476 with an expiration date of May 31, 2006. If you have comments regarding the burden estimate or any other aspect of this collection of information, you may send them to : NIH Project Clearance Branch, 6705 Rockledge Drive, MSC 7974, Bethesda, MD 20892-7974, ATTN: PRA 0925-0476.

Before beginning, please feel free to print these instructions using your browser's print feature.

If you have questions about this survey, please send email to ghreval@llhc.nlm.nih.gov

Explore Genetics Home Reference

Please explore the Genetics Home Reference site. Please read any content of interest; we encourage you to take a look at the area that contains information about conditions and genes.

After exploring the site, use the link in the top-right corner of any page to return to this survey and proceed to the questionnaire.

In one sitting, please answer all the questions and click the submit button at the bottom of the form. We will receive your responses only after you click on the submit button.

We appreciate and value your opinion. Please be confident that your anonymity and confidential answers will be protected at all times.

Click here to explore Genetics Home Reference

Click here to complete the questionnaire

GHR Questionnaire

In one sitting, please answer all the questions and click the submit button at the bottom of the form. We will receive your responses only after you click on the submit button.

1. Please assess the importance of the following five features of the Genetics Home Reference web site. (Click one circle for each feature, please).

Search

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

Browse conditions by category

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

Browse genes by category

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

Help Me Understand Genetics

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

Resources and patient support

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

2. When reviewing conditions at the site, how important to you were the following features

The 'What is the condition' section?

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

Genetic causes of the condition

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

Inheritance pattern

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

Links to additional information

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

3. From a condition description page, how easy was it to find the link to related genes?

Very Easy Easy Neither easy or difficult Difficult Very difficult Could not find Did not review

4. When reviewing genes at the site, how important to you were the following features:

Normal function of the gene

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

Conditions associated with mutations in the gene

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

Links to additional information

Very important Important Neither important or unimportant Unimportant Very unimportant Could not find Did not review

5. From a gene description page, how easy was it to find the link to related conditions?

Very Easy Easy Neither easy or difficult Difficult Very difficult Could not find Did not review

6. Was it easy or difficult to find the information you were seeking? (one response, please, place an X to the left of the answer you select)

Very easy Easy Neither easy or difficult Difficult Very difficult

7. Was it easy or difficult to move from the Genetic Home Reference homepage to other areas within the site? (one response, please, place an X to the left of the answer you select)

Very easy Easy Neither easy or difficult Difficult Very difficult

8. Was it easy or difficult to return to previously viewed pages? (one response, please, place an X to the left of the answer you select)

Very easy Easy Neither easy or difficult Difficult Very difficult

9. Was it easy or difficult to use the search box? (one response, please, place an X to the left of the answer you select)

Very easy Easy Neither easy or difficult Difficult Very difficult

10. When you searched on the site or clicked on links, how quickly did the site respond? (one response, please, place an X to the left of the answer you select)

Very fast Fast Neither fast or slow Slow Very slow

11. Overall, do you find it easy or difficult to read information about genetics from a computer compared to a print source, such as a book or pamphlet? (one response, please, place an X to the left of the answer you select)

Very easy Easy Neither easy or difficult Difficult Very difficult

12. Do you find the Genetics Home Reference web site well or poorly designed? (one response, please, place an X to the left of the answer you select)

Very well designed Well designed Neither well or poorly designed Poorly designed Very poorly designed

13. Overall, was the text on the site easy or difficult to read? (one response, please, place an X to the left of the answer you select)

Very easy Easy Neither easy or difficult Difficult Very difficult

14. In what role did you visit the Genetics Home Reference today? (Check all that apply, please)

__ Patient with Specific Condition
__ Family or Friend of Patient
__ Student (college/graduate school)
__ Student (grades 7-12)
__ General Health Consumer
__ Health Care Provider
__ Genetics Professional
__ Other Researcher or Scientist
__ Educator

15. What is your overall satisfaction with the Genetics Home Reference? (one response, please, place an X to the left of the answer you select)

Very dissatisfied Dissatisfied Neither satisfied or dissatisfied Satisfied Very satisfied

16. Did you find information about the genetic condition that is most important to you in Genetics Home Reference today? (one response, please, place an X to the left of the answer you select)

Yes No Don't know

17. Will you recommend the Genetics Homes Reference web site to someone else? (one response, please, place an X to the left of the answer you select)

Yes Maybe No Don't know

18. Will you return to the Genetics Home Reference web site in the future? (one response, please, place an X to the left of the answer you select)

Yes Maybe No Don't know

19. What outcomes do you think may result from visiting the Genetics Home Reference web site? (Check all that apply, please):

___ Will improve my ability to understand a genetics professional explain a genetic condition
____ Will improve my understanding of a genetic condition
___ Will improve my ability to assist as a caregiver
___ Will consider looking for more health information
___ Will seek information from a library
___ Will consider joining a local group with common interest in a genetic condition
___ Will consider joining an on-line users group with an interest in a genetic condition
___ Other
___ Nothing specific will happen

20. Below is a list of paired, opposite words that describe feelings and impressions you may have after using the Genetics Home Reference website. Please tell us how you feel, or your general impressions about the Genetics Home Reference web site.

Here's a guide to respond to all the questions in this section: In the first question, for example, if check the box next to Authoritative, then, you believe the Genetics Home References is very authoritative, if you check the box next to Unreliable, then, you find the Genetics Home Reference is very unreliable. If you check in the middle, then, you find the Genetics Home Reference is neither authoritative nor reliable. Please follow the same pattern through all the answers; there is no 'right' answer -- just tell us what you think!

The Genetics Home Reference Website strikes me as: *(Please mark one answer for each pair)*

Authoritative ____: ____: ____: ____: ____: ____: ____: Unreliable

Valuable ____: ____: ____: ____: ____: ____: ____: Worthless

Complex ____: ____: ____: ____: ____: ____: ____: Simple

Appealing ____: ____: ____: ____: ____: ____: ____: Unappealing

Accessible ____: ____: ____: ____: ____: ____: ____: Inaccessible

Pleasant ____: ____: ____: ____: ____: ____: ____: Unpleasant

Accurate ____: ____: ____: ____: ____: ____: ____: Inaccurate

Well-designed ____: ____: ____: ____: ____: ____: ____: Poorly designed

Biased ____: ____: ____: ____: ____: ____: ____: Unbiased

Pertinent ____: ____: ____: ____: ____: ____: ____: Not pertinent

Up to date ____: ____: ____: ____: ____: ____: ____: Outdated

Informative ____: ____: ____: ____: ____: ____: ____: Uninformative

Readable ____: ____: ____: ____: ____: ____: ____: Unreadable

Messy ____: ____: ____: ____: ____: ____: ____: Neat

Familiar ____: ____: ____: ____: ____: ____: ____: Unfamiliar

Friendly ____: ____: ____: ____: ____: ____: ____: Unfriendly

Inspiring ____: ____: ____: ____: ____: ____: ____: Uninspiring

Like ____: ____: ____: ____: ____: ____: ____: Dislike

21. Before visiting the Genetics Home Reference today, how would you describe your interest in learning about genetic disorders and conditions? (one response, please, place an X to the left of the answer you select)

Very interested Interested Neither Interested or Uninterested Uninterested Very uninterested

22. Before visiting the Genetics Home Reference today, how would you describe your knowledge about genetic disorders and conditions? (one response, please, place an X to the left of the answer you select)

Well informed Informed Neither informed or uninformed Uninformed Very uninformed

23. Please select the category that includes your age. (one response, please, place an X to the left of the answer you select)

24 and under
25-34
35-44
45-54
55-64
65 and over

24. What is your Gender? (one response, please, place an X to the left of the answer you select)

Female
Male

25. Which of the following best describes the highest level of education you have completed? (one response, please, place an X to the left of the answer you select)

Did not complete high school
High school or equivalent graduate
Some college /vocational school
College graduate
Some postgraduate school
Graduate/professional degree

26. Which of the following best describes the area you live in? (one response, please, place an X to the left of the answer you select)

Urban
Suburban
Rural

27. On average, how many hours a day (both at home and at work) do you check for email as well as surf the web? (one response, please, place an X to the left of the answer you select)

0 to 1 hours
2 to 4 hours
5 to 6 hours
More than 6 hours

28. What type of computer did you use to tour Genetics Home Reference today? (one response, please, place an X to the left of the answer you select)

PC
Macintosh
Other
Don't know

29. How did you access the internet today (one response, please, place an X to the left of the answer you select)

Through a telephone modem
Through a cable modem or DSL
Through a wireless connection
Through a T1, or high speed line
Other
Don't know

APPENDIX B: CURRICULUM VITAE

Joyce A. Mitchell, PhD
Robert A. Logan, PhD

Content Developers
- Sherri C. Calvo, MS, MBA
- Cathy Fomous, PhD
- Stephanie Morrison
- Diane Mucci, PhD

System Developers
- Jane Fun (Lead)
- Phillips Wolf

System Evaluation
- May Cheh, MS

Joyce A. Mitchell **Professor and Chair, Department of Medical Informatics**

Education and Training

Stephens College	BA	1972	Mathematics & Biology
University of Wisconsin	PhD	1976	Population Genetics
University of Missouri-Columbia	Postdoc	1978	Medical Information Science
University of California, San Francisco	Postdoc	1980	Medical Genetics

Research and Professional Experience:

Academic Positions

2005 - present Chair and Professor, Dept of Medical Informatics, School of Medicine, University of Utah, Salt Lake City, UT

2003 - 2005 Adjunct Professor, School of Information Science and Learning Technology, College of Education, University of Missouri-Columbia (UMC), Columbia, MO

2001- 2003 Senior Scholar, Lister Hill National Center for Biomedical Communications, National Library of Medicine, Bethesda, MD

1998 - 2005 Professor, Dept of Health Management and Informatics, UMC, Columbia, MO

1992 - 2005 Professor, Dept of Child Health (Medical Genetics), School of Medicine, UMC Columbia, MO

1990 - 2000 Adjunct (Associate/Full) Professor, Computer Science, Dept of Computer Engineering and Computer Science, College of Engineering, UMC, Columbia, MO

1986 - 1992 Associate Professor, Dept of Child Health, School of Medicine, UMC, Columbia, MO

1980 - 1986 Assistant Professor, Dept of Child Health (Medical Genetics), School of Medicine, UMC, Columbia, MO

Administrative Positions

2003 - 2005 UMC Campus Coordinator for Bioinformatics, Columbia, MO

2002 - 2005 Director, Biomedical and Health Informatics Training Program, UMC, Columbia, MO

2002 - 2005 Interim Director, Health Informatics, UMC, Columbia, MO

1994 - 2001 Chief Information Officer, University of Missouri Health Care, Columbia, MO

1994 - 2001 Associate Dean, Integrated Technology Services, School of Medicine, UMC, Columbia, MO

1989 - 1998 Director, Medical Informatics Group, UMC, Columbia, MO

1984 - 1989 Director, Information Sciences Group, UMC, Columbia, MO

Other Experience and Professional Memberships

Board Certification: American Board of Medical Genetics – Ph.D. Medical Geneticist

Professional Organization Memberships:

American Medical Informatics Association (AMIA)
American College of Medical Informatics (elected Fellow)
College of Healthcare Information Management Executives (CHIME)
Health Care Information Management Systems Society (HIMSS)
American College of Medical Genetics (ACMG), Founding Fellow

1991-92; 95-96; 2005-08 Elected to Board of Governors, American College of Medical Informatics

1999 - 2001 Elected to Executive Governing Board, CIO Council of University Healthcare Consortium (UHC)

1994 - 2003 Elected to Board of Governors, Integrated Advanced Information Management Systems (IAIMS) Consortium

1982 - 2005 Member, Governor's Advisory Council on Genetic Diseases, State of Missouri

Honors

1998	Ida and George Eliot Prize (best paper), Medical Library Association
1988 - 1989	Chair, Biomedical Library Review Panel (NIH study section), NLM
1996 - 1997	Chair, Board of Scientific Counselors for Intramural Research, NLM

Selected peer-reviewed publications (partial listing since 2001, in chronological order)**:**

Lobenstein KW, Mitchell J, Hodge R. Taking telemedicine into the mainstream. NLM Symposium on Telemedicine and Telecommunications: Options for the New Century, March 2001. www.nlm.nih.gov/research/telesymp.html.

Srinivasan P, Mitchell JA, Bodenreider O, Pant G, Menczer F. Web crawling agents for retrieving biomedical information. Proceedings of the NETTAB Conference on Agents in Bioinformatics, July 12-14, 2002 and also the Proceedings of the Symposium on Bioinformatics and Multi-Agent Systems (BIXMAS). July 15, 2002. Bologna, Italy.

Mitchell JA, McCray AT, Bodenreider O. From Phenotype to Genotype: Experiences in navigating the available information resources. J Am Med Inform Assoc suppl 2002:1109.

Bodenreider O, Mitchell JA, McCray AT. Evaluation of the UMLS as a terminology and knowledge resource for biomedical informatics. J Am Med Inform Assoc suppl 2002:61-65.

Hicks LL, Boles KE, Hudson S, Kling B, Tracy J, Mitchell J, Webb W. Patient satisfaction with teledermatology services. J of Telemed Telecare. 2003;9(1):42-5.

Bodenreider O, Mitchell JA, McCray AT. Biomedical ontologies. Proceedings of the Pacific Symposium on Biocomputing, 2003:562-564 and 2004:164-165.

Bodenreider O, Burgun A, Mitchell JA. Evaluation of WordNet as a source of lay knowledge for molecular biology and genetic diseases: a feasibility study. Stud Health Technol Inform 2003;95:379-84.

Hristovski D, Peterlin B, Mitchell JA, Humphrey SM. Improving literature based discovery support by genetic knowledge integration. Stud Health Technol Inform 2003;95:68-73.

Mitchell, JA, McCray AT, Bodenreider O. From phenotype to genotype: issues in navigating the available information resources. Methods Inf Med. 2003;42(5):557-63.

Mitchell JA, Aronson AR, Mork JG, Folk LC, Humphrey SM, Ward JM. Gene Indexing: characterization and analysis of NLM's GeneRIFs. AMIA Annu Symp Proc 2003:460-4.

Pancoast PE, Patrick TB, Mitchell JA. Physician PDA use and the HIPAA Privacy Rule. J Am Med Inform Assoc. 2003;10(6):611-2.

Patrick TB, Demiris G, Folk LC, Moxley DE, Mitchell JA, Tao D. Evidence-Based Retrieval in Evidence-Based Medicine. J Med Libr Assoc. 2004 Apr;92(2):196-9.

Johnson ED, Pancoast PE, Mitchell JA, Shyu CR. Design and evaluation of a personal digital assistant-based alerting service for clinicians. J Med Libr Assoc. 2004 Oct;92(4):438-44.

Demiris G, Patrick TB, Mitchell JA, Waldren SE. To telemedically err is human. Jt Comm JQual Saf. 2004 Sep; 30(9):521-7.

Mitchell JA, Fun J, McCray AT. Design of the Genetics Home Reference: a new NLM consumer health resource. J Am Med Inform Assoc. 2004 Nov-Dec; 11(6):439-47.

Mitchell JA. The impact of genomics on e-health. In: Demiris G (ed). e-Health: Current Status and Future Trends, vol 106: Studies in Health Technology and Informatics. Amsterdam: IOS Press, 2004. 63-74.

Hristovski D, Peterlin B, Mitchell JA, Humphrey SM. Using literature-based discovery to predict disease candidate genes. Int J Med Inform 2005 Mar;74(2-4):289-98.

Popescu M, Keller J, Mitchell JA. Fuzzy measures on the Gene Ontology for gene product similarity. Trans of the IEEE on Comput Biol and Bioinf, 2005 (in press).

Mitchell JA, Demiris G. Telegenetics: the next phase in the provision of genetic services? Genet Med, 2005: Jan;7(1):1-2.

Robert A. Logan

Social science analyst; Senior scholar; Director, Informatics Training

Education and Training

Tulane University	B.A.	1969	History
University of Missouri	M.A.	1973	Journalism
University of Iowa	Ph.D.	1977	Mass Communication

Research and Professional Experience:

Social Science Analyst, Program Director, Informatics Training, Lister Hill National Center for Biomedical Communication; Senior staff, National Library of Medicine, National Institutes of Health. (2003-present)

Research specialty areas: public understanding of science and medicine; evaluation of consumer health informatics; consumer health informatics outreach; Q technique and methodology; news media content analysis and ethics within the major professions.

Professor Emeritus, University of Missouri-Columbia. (2003-present)

Associate Dean, Professor, Director, Science Journalism Center, School of Journalism, University of Missouri-Columbia. (1986-2003)

Member Graduate & Ph.D. Faculty. Administered undergraduate studies plus a privately funded program to a) improve the practice of science journalism and b) foster research regarding the public understanding of science and medicine. Taught graduate news media and society, social science research methods and science writing courses. Tenured, June 1988. Promoted to full professor, August 1993. Associate Dean for Undergraduate Studies, August 1993 - January, 2003. Took voluntary early retirement on January 1, 2003. Appointed Professor Emeritus January 15, 2003.

Recent Honors:

National Library of Medicine's Director's award for: Native American Listening Circles, Information prescription program and Evaluation of MedlinePlus (2004)

External examiner. School of Journalism and Mass Communication, Nanyang Technological University, Singapore (2002-2003)

Distinguished Lecturer, National Journalism Training Organization, Auckland, New Zealand (2000 and 2003)

Member, Science Communication Program Review Board, Vanderbilt University (1999-2002)

Missouri Arthritis Rehabilitation Research and Training Center web site. Site received Medinex and Health Way citations plus other national awards for public communication of medicine (1999)

Member, Research and Roadmap for Public Understanding of Science in the 21st Century, a national board coordinated by NASA-Marshall Space Center (1998 – 2002)

Member, The New York *Times* College Advisory Board (1997 – 2001)

Refereed Publications (since 2000)**:**

Book Chapters

Logan, Robert A; MacLean Malcolm Jr; Stephenson, William: Career contributions and leadership. In: L. Manca and G.W. Pieper (eds.). A heretic in American journalism education and research: Malcolm S. MacLean Jr. Revisited. Columbia, MO: Stephenson Research Center, 2001, p. 215-226.

Articles and Conference Proceedings

Logan RA, Park J, Shin JH. Elite sources, context, and news topics: how two Korean newspapers covered a public health crisis. Sci Commun. 2004 June; 25(4):365-89.

Logan RA, Shin JH, Park J. Prevailing impressions of social actors in Korean news coverage of a public health crisis. Sci Commun. 2004 June; 25(4):399-418.

Logan RA. Evaluating consumer informatics: learning from health campaign research. Medinfo. 2004; 2004:1147-51.

Gemoets D, Rosemblat G, Tse T, Logan RA. Assessing readability of consumer health information: an exploratory study. Medinfo. 2004;2004:869-73.

Robert A. Logan, Stephenson, MacLean & Qualitative Mass Communication Research. Journal of Human Subjectivity 2003;1(2):4-30.

Caburnay CA, Kreuter MW, Luke DA, Logan RA, Jacobsen HA, Reddy VC, Vempaty AR, Zayed HR. The news on health behaviors: coverage of diet, activity, and tobacco in local newspapers. Health Educ Behav. 2003 Dec;30(6):709-22.

Motavalli P, Patton M, Logan RA, Frey C. Promoting environmental writing in undergraduate soil science programs. J Nat Resour Life Sci Educ. 2003;32:93-99.

Logan RA, News' compartmentalization: implications for food biotechnology coverage. AgBioForum. 2002;4(3&4):194-198.

Logan RA, Shibuya A. Sustaining and challenging cultural norms: Yomiuri's & Asahi's coverage of full cancer disclosure in the 1990s. Keio Communication Review. 2002; 24:71-92.

Logan RA. Science mass communication: its conceptual history. Sci Commun. 2001;23(2):135-63.

Logan RA, Nuttall RJ, Hazelwood SE, Parker JC, Johnson JC, Hewett JE, Reid JC. Audience motivations to use an arthritis website. Arthritis Care Res. 2000 Oct;13(5):320-9.

Logan RA, Zengjun P, Wilson NF. Science and medical coverage in the Los Angeles Times and the Washington Post: a six year perspective. Sci Commun. 2000;22(1):5-26.

Logan RA, Zengjun P, Wilson NF. Prevailing impressions in science and medical news: a content analysis of the Los Angeles Times and the Washington Post. Sci Commun. 2000;22(1):27-45.

Articles under current review: 4

Sherri C. Calvo — **Genetics Home Reference Content Developer**

Education and Training

Rutgers University	BA	1980	Physics
University of Maryland	BS	1999	Astronomy
New York University	MBA	1984	Computer Applications and Information Systems
Johns Hopkins University	MS	2004	Biotechnology

Research and Professional Experience:

Information Research Specialist, National Library of Medicine, National Institutes of Health (2002-present)

Member of the Genetics Home Reference content development team. Assist with content issues for other medical informatics projects including Clinical Questions and Profiles in Science.

Senior Scientist, Global Science and Technology (2001-2002)

Served on a NASA-sponsored research team studying emerging intelligent scientific information systems technologies, with the goal of positioning the Federal government to take maximum advantage of these technologies and make the best possible use of available funding.

Science and Medical Writer, freelance (1999-2002)

Assignments included online content for GenomeWeb, BioMedNet, and ChemWeb, articles for the magazines Genome Technology and BioTechniques and the newsletter BioInform, and material for the Encyclopedia of Technology and Applied Science (Marshall Cavendish, 1999), Science and its Times (Gale Group, 2000), Science in Dispute (Gale Group, 2001), World of Genetics (Gale Group, 2001) and other reference works.

Computer Scientist, NASA Goddard Space Flight Center (1991-1999)

Technical director for the High Energy Astrophysics Science Archive Research Center (HEASARC), leading the development of web applications for data access and a "meta-information" database to allow querying heterogeneous, distributed digital collections. Lead systems engineer for the EOSDIS science user interface. Instrumental in developing the successful EOSDIS prototype, which allowed simultaneous query of heterogeneous distributed scientific information systems at multiple data centers.

Senior Scientific Systems Analyst, STX Corporation (1990-1991)

Served on the design team for NASA's EOSDIS Version 0 prototype. Worked with scientists to define requirements, metadata and interfaces for the system. Surveyed user interfaces of numerous scientific data systems for their strengths and weaknesses in order to develop specifications for the prototype. Researched and evaluated technologies for implementation.

Technical Writer, Laboratory Assistant, Unisys Corporation (1989-1990)
Wrote laboratory reports for NASA's Parts Analysis Department. Examined electronic components using a variety of techniques including electron microscopy, mass spectrometry and radiography.

Honors:
National Institutes of Health Staff Recognition Awards (2003, 2004)
Space Act Innovation Award (Group award for EOSDIS prototype, 1996)
National Performance Review Silver Hammer Award (Group award for EOSDIS prototype, 1994)
Other NASA individual and group awards (12 awards, 1991-1999)

Publications:
Ramapriyan HK, Kempler S, Lynnes C, McDonald KR, McConaughy G, Kiang R, Calvo SC, L. Roelofs L, Harberts R, Dun D. Conceptual study of intelligent data archives of the future. Proceedings of the IEEE Symposium on Mass Storage Systems, 2002.

Calvo SC, White NE, McGlynn TA, Duesterhaus MM, Rosen CA, Sabol EJ. An enriched meta-information schema for astronomical databases. Proceedings of the Conference on Astronomical Data Analysis and Software Systems, 1995.

Calvo SC, White NE, McGlynn TA, Yom SH. Meta-information in the next-generation HEASARC database. Bulletin of the American Astronomical Society, May 1995.

Calvo SC, McDonald KR. Accessing distributed heterogeneous Earth science inventories via the EOSDIS Version 0 Information Management System. Proceedings of the Workshop on Intelligent Access to On-Line Digital Libraries, IEEE Conference on Artificial Intelligence and Applications, 1994.

McDonald KR, Calvo SC. Accessing Earth Science Data from the EOS Data and Information System. Proceedings of the Goddard Conference on Mass Storage Systems and Technologies, 1992.

Cathy Fomous **Genetics Home Reference Content Developer**

Education and Training

University of New Hampshire	B.S.	1974	Botany
Texas Tech University	M.S.	1981	Botany
Georgetown University	Ph.D.	1988	Genetics

Certification:

Genetic Counselor, American Board of Medical Genetics (issued 1987)
Teacher Certification, State of Virginia (issued 1989), State of Texas (issued 1976)

Research and Professional Experience:

Scientist IV, Aspen Systems Corporation (2002-present)

Senior scientist for Genetics Home Reference. Evaluate biomedical literature for web site content and structure. Analyze information from genetic databases (e.g., Entrez Gene, SwissProt) for content development. Translate complex biomedical information to user-friendly language and format. Interact with genetics experts for review of web site content. Work with team members to enhance web site design and navigation

Vice President, Scientific Research Evaluation, Council for Responsible Nutrition (CRN) (1999-2002)

Developed research strategy for CRN membership. Initiated and developed partnerships with government agencies, trade associations, and scientific experts to address issues related to safety and efficacy of vitamin and mineral supplements. Provided expertise in genetics, genetic toxicology, and gene-nutrient interactions. Assisted membership with regulatory questions related to the Dietary Supplement Health and Education Act (DSHEA).

Part-time Faculty Appointment, George Washington University (1997-2000)

Developed curriculum for a graduate cytogenetics course that integrated classical cytogenetics with current advances in molecular genetics techniques and protein/nucleic acid biochemistry.

Science Writer, Tascon, Inc. (1997-1999)

Provided analytical and technical support to biomedical and health agencies. Collected, synthesized, and organized complex biomedical information for audiences with diverse levels of understanding. Interpreted salient results of biomedical research and communicated this information in written materials appropriate for healthcare professionals and the general public.

Editor, Paragraphics (1996-1997)

Edited science textbooks for accuracy and pedagogy. Edited and/or wrote teacher's edition with a focus on improving critical thinking and problem solving skills, meeting diverse needs of students, and developing multicultural perspectives.

Project Leader, Institute of Toxicology, German Research Center for Nutrition (1993-1994)
Directed experiments in cancer studies using immunocytochemical techniques for detection of the proliferation cell nuclear antigen (PCNA) and a second proliferation marker (Ki67). Studied effects of genotoxic treatment on the proliferation index *in vitro* and *in vivo.*

Director, Cytogenetics Laboratory, Columbia Hospital for Women (1988-1989)
Directed laboratory analyzing blood and amniotic fluid samples for chromosome abnormalities. Evaluated and signed out genetic diagnostic reports. Counseled physicians and patients concerning diagnostic reports and patient risk.

Genetic Counselor, Georgetown University (1984-1988)
Counseled patients at risk for genetic disorders or chromosomal abnormalities. Communicated with referring physicians outlining information provided to patients. Answered requests for genetic information from patients and physicians.

Predoctoral Student, Biology Department, Georgetown University (1981-1988)
Conducted research in genetic toxicology comparing the response to DNA damage in three fetal cell types. Research led to a further understanding of the biological properties of amniotic fluid cell types and their optimal use in antenatal diagnosis.

Electron Microscope Technician, Anatomy Department, Texas Tech University School of Medicine (1979-1981)
Managed a laboratory with research grants in the areas of neural crest development and diabetes research. Responsible for general laboratory duties related to electron microscopy.

Other (1976-1988)
More than 12 years experience teaching on several levels—high school, undergraduate, graduate, medical school, and continuing education for health professionals. Subject matter included biology, chemistry, physics, and genetics.

Publications:

Fomous CM, Costello RB, Coates PM. Symposium: conference on the science and policy of performance-enhancing products. Med Sci Sports Exerc. 2002;34(10):1685-90.

Fomous CM, Cardellina JH 2nd. St. John's wort and major depression. JAMA. 2001;286(1):42.

Cardellina JH II, Fomous C. Your genome—just another credit card to carry around. Fertil Steril. 1999;72(2):378-9.

Stephanie Morrison **Genetics Home Reference Content Developer**

Education and Training

College of William and Mary	B.S.	1999	Biology (major), Anthropology (minor)
Medical College of Virginia	graduate study	1999-2000	Genetic Counseling

Research and Professional Experience:

Writer/Editor, Aspen Systems Corporation, Genetics Home Reference Project (2002-present)

Research write, and edit summaries of genetic disorders, genes, and chromosomes for the Genetics Home Reference, an online resource from the National Library of Medicine. Develop and update a multi-chapter genetics primer for the Genetics Home Reference. This resource explains the basics of genetics, hereditary disorders, and the Human Genome Project in consumer-friendly language. Collaborate with medical illustrators to create illustrations for the primer. Work closely with web developers to design and implement new technical features of the web site. Maintain the team's internal web site, including a style guide for the Genetics Home Reference's written content.

Writing Coordinator, Aspen Systems Corporation, Cancer Information Service Central Support (2001-2002)

Tracked and reviewed the writing team's many assignments and served as a mentor to staff. Researched, wrote, and edited National Cancer Institute fact sheets and public correspondence. Developed and implemented task training for new writing team staff. Compiled written and statistical information about staff activities for monthly client reports.

Cancer Information Writer, Aspen Systems Corporation, Cancer Information Service Central Support (2000-2001)

Researched, wrote, and edited National Cancer Institute fact sheets and public correspondence. Prepared cancer-related proclamations, briefings, and greetings on behalf of the National Cancer Institute, the White House, and other federal government agencies. Researched and presented information on current cancer topics at bi-monthly staff seminars.

Honors:
Nominated as an Aspen Extraordinary Employee, 2001.
Inducted into the Phi Beta Kappa Society, 1999.

Publications:
Patt J, Morrison S. National Cancer Institute resources for patients and their caregivers. Cancer Practice. 2001;9(5):257-61.

Diane Mucci **Genetics Home Reference Content Developer**

Education and Training

College of Mount St. Joseph	B.S.	1989	Biology
University of Cincinnati College of Medicine	Ph.D.	1995	Molecular Genetics, Biochemistry, and Microbiology

Research and Professional Experience:

Scientist IV, Aspen Systems Corporation (2000-present)

Develop, implement, and disseminate information products produced by National Library of Medicine for the Genetics Home Reference web site, a consumer friendly web site with information about genetic disorders and the genes that cause them. Major responsibilities include technical writing, data and literature evaluation, and revising and updating databases and web site review and update.

Associate Professor of Biology, Northern Virginia Community College (2004-Present)

Teach courses in general biology, anatomy and physiology, microbiology, and biotechnology. Assist in course and curriculum development for the biotechnology degree and certificate programs. Develop community contacts and create advisory board for biotechnology program oversight. Assist in outfitting new laboratories with equipment and staff to increase course offerings in biotechnology and allied health care-related fields.

Assistant Professor of Microbiology/Bioscience Laboratory Technician Program Manager, Frederick Community College (2000-2004)

Developed and implemented curriculum for the BLT associate degree program and certificate program; coordinated contacts and program support with local biotech industries; held advisory board meetings; taught courses in microbiology, biotechnology, forensic biology, and genetics.

Postdoctoral Fellow, Food and Drug Administration Center for Biologics Evaluation and Research at the National Institutes of Health (1995-1998)

Performed laboratory research in classical and molecular genetics, specifically studying gene expression, gene regulation, and gene targeting in *Drosophila melangaster*.

Honors:

Frederick Community College – Executive Leadership Program 2003
Frederick Community College – President Faculty Association 2003-2004

Publications:

Brown JL, Mucci D, Whiteley M, Dirksen ML, Kassis JA. The Drosophila Polycomb group gene pleiohomeotic encodes a DNA binding protein with homology to the transcription factor YY1. Mol Cell. 1998 Jun;1(7):1057-64.

Mucci D, Forristal J, Strickland D, Morris R, Fitzgerald D, Saelinger CB. Level of receptor-associated protein moderates cellular susceptibility to pseudomonas exotoxin A. Infect Immun. 1995 Aug;63(8):2912-8.

Jane Fun **Genetics Home Reference System Development Lead**

Education and Training

Pennsylvania State University	B.S	1985	Electrical Engineering
Syracuse University	graduate study	1986-1988	Electrical Engineering

Research and Professional Experience:

Senior System Architect, Thoughtful Solutions, Inc. (2002-present)

Design Genetics Home Reference system architecture. Evaluate genetic and medical research databases and create methods to extract and apply data. Evaluate and integrate available software services, frameworks, and techniques. Write and edit project descriptions and papers. Lead team to achieve research, development, and maintenance objectives.

Software Development Lead, GEICO, Inc. (2001-2002)

Managed team to deliver workflow components to support automated processing of medical bills related to claims processing. Designed object and data models. Wrote C++, XSL, and SQL scripts. Integrated and tested components including VisualBasic and C++ on NT, C++ and Oracle on AIX, and COBOL on IBM mainframe.

Project Manager, Foundry, Inc. (2000-2001)

Managed team to deliver a web site that provided approval for refinancing mortgages for the Chase Manhattan Mortgage Company. The site included both customer and administrative portals. Defined requirements, developed and managed project plan, wrote Java and JSP code, and coordinated testing and deployment. Technology for the project included Weblogic, Oracle, Java, XML, XSL, and a custom JSP-like layer.

Java Developer, Foundry, Inc. (2000)

Developed Java servlets for a web-based sports retailer. Also installed and customized Bugzilla bug-tracking software. Used Apache, Weblogic, Oracle, JavaBeans, EJB, and JSP technologies.

Senior System Architect, AppNet, Inc. (1999-2000)

Designed and developed administrative support systems for an e-commerce shopping mall. Led team, consulted on system development processes, and provided system support. Designed and implemented processes to collect catalog, promotion, and inventory data before automation was available. Designed transition strategies between manual and automated systems. Developed both graphical and electronic interfaces to support mall-owner, merchant, and fulfillment users. Also developed reporting requirements and reports. Technology included NES, JSP, EJB, Weblogic, TopLink, Oracle, EDI, XML, and Brio Enterprise technologies.

System Architect, AppNet, Inc. and Lockheed Martin Corp. (1998)

Consulted on a project to migrate an automotive insurance claim's processing system from a 1-tier mainframe to a 3-tier client-server architecture. The system inserted a VisualBasic GUI client, a C++ application server, and a persistence server using Oracle

in front of legacy mainframe COBOL applications. Mentored staff and wrote requirements as Use Cases and Line Item Requirements. Advised on team organization, scheduling, risk assessment, software re-use, configuration management, and the Rational Unified Process. Designed and led team to develop the core claim's processing subsystem. This included coding a large part of the application server in C++ using Microsoft Visual Studio and the Rogue Wave Tools.h++. Also designed an XML-like pattern used to communicate data between components.

Instructor and Senior Consultant, Lockheed Martin Advanced Concepts Center (1997)
Taught and developed seminar-style courses on the technical and management aspects of distributed computing using object-oriented analysis and design, client-server technology, project management and testing.

Technical Lead/Project Manager, Lockheed Martin Advanced Concepts Center (1996-1997)
Led project to develop an order-entry system for reselling local phone service. Developed the initial system concept, gathered and managed requirements, helped implement, test, install, and maintain the system. The system had a 2-tier architecture with Objective-C clients over an Oracle database server. Client and server sites were distributed across five states. The system also had interfaces with a mainframe-based billing and a PC-based customer care applications. The system provided electronic interfaces between the service originators and reseller in EDI and other formats.

Project Manager and Systems Engineer, TASC, Inc. (1989-1996)
Performed a wide variety of tasks, moving from developing and using software tools to managing software development teams. Performed analysis and developed algorithms to determine capacity and maximize efficiency of a large communication system.

Engineer, General Electric Co. (1985-1989)
Developed and operated mainframe-based sensor simulators. Evaluated designs for radars, antennas, target tracking, and infrared systems. Designed digital hardware for a phased array antenna.

Honors:
TASC Special Achievement Award, 1995
Lockheed Martin President's Award, 1997

Publications:
Mitchell JA, Fun J, McCray AT. Design of the Genetics Home Reference: a new NLM consumer health resource. J Am Med Inform Assoc. 2004;11(6):439-47.

Phillips Wolf **Genetics Home Reference System Developer**

Education and Training

Grinnell College	B.A.	1990	Russian Language and Literature

Research and Professional Experience:

Systems Architect, Aquilent, Inc., 1998-present:

Genetics Home Reference web site at the National Library of Medicine

User interface, web server Java programming using Turbine/Velocity, automated content creation using Perl, Java, XSL, and MySQL, automated conversion of word-processor documents to XML, HTML, and PDF.

NAVSEA "SeaPort" procurement web-portal

Users spin task orders off a multi-vendor reusable "IDIQ" contract using a wizard and a library of past experience, then follow the procurement through bid evaluation, award, and performance. Use cases, test plans; coordination of programmers, database architecture, software design, and web page design; programming in SQL, Active Server Pages, Visual Basic, C++, and Perl.

Commercial B2B engineered-parts procurement portal

Users build complex RFPs from a library of templates, then track them through bidding, award, and fulfillment. Features for content creation, publishing, invoicing, and "I Agree" downloads; distributed concurrent publishing system using Java RMI, JNI, and Perl; coordination of subsystem programmers; XML processing in Java (SAX, DOM and XSLT); customization of BladeRunner Content Creator (from BroadVision); Microsoft Word macro templates for content validation.

Senior Software Engineer, Aquilent, Inc., 1997-1998:

Fingerprint Workstation

Users scan fingerprint "tenprint" cards; the software submits the scans to databases for automated comparison; users evaluate the "hits" and manipulate prints to enhance or sketch ridges and features. TWAIN scanning, storage, business logic, and interface to a very remote server; DCOM architecture allowing on-site customization of the user interface; queuing architecture allowing work to continue even when the server is off-line; GUI widgets that populate asynchronously from remote back-end server; multithreading to stay "live" while the cantankerous back-end seizes up; test tool for stress and regression (in Perl); automated predelivery build scripts.

Software Engineer, Aquilent, Inc., 1993-1996:

Local telephone business software performance appraisal

Instrumented and stressed CORBA-based Objective-C and C++ systems

Home Banking Software

Users enjoyed banking services via modem, checkbook register window & custom GUI widgets (Windows/C++); statement reconciliation feature.

INTELSAT Business Systems Integration & Test

Demonstration project of application of automated testing to booking-and-billing systems in Gupta SQLWindows and C++, in Windows and OS/2. Responsible for designing and writing test scripts; assessed applicability of automated testing to Windows client-server 4GL clients.

Cash Register Device Drivers for Windows

Responsibilities included design an API for control of the cash drawer, card swiper, and LCD screen; device drivers for MS-DOS and Windows 3.1 protected-mode (VxD) and standard-mode; programmer documentation.

Dial-up Online System User Interface

It replaced a plaintext terminal user interface for file downloads, chatting, game playing, etc. (Windows/C++/MFC/ODBC); modem-detection, connection-management, and database subsystems; customer service to the online service's help desk.

NASA Software-Requirements System

Users built requirements documents; user interface pages (in "Omnis" cross-platform Windows/Mac tool); performance optimization by writing C-language DLLs/code resources; Sybase SQL.

Programmer/Analyst, The Rochester Group, Inc., 1991-1993:

Multidimensional Business-Forecast Tool

Users cross-tabulated and analyzed transactions using a multi-dimensional query and formula language. Windows 3.0; in C++ using XVT portability tool; user interface for visualizing multi-dimensional data; multidimensional data store.

May Cheh **Computer Scientist, Computer Science Branch, LHNCBC**

Education and Training

Univ of California, Berkeley	B.A.	1970	Chemistry (major), Mathematics (minor)
American University, Washington DC	M.S.	1983	Computer Science

Certification:
Secondary Education Certification, State of California (issued 1971)

Research and Professional Experience:

Computer Scientist, Computer Science Branch, LHNCBC (1980-present)
Design, develop and coordinate LHNCBC's intramural training programs, including its fellows program, medical student elective, NLM rotation for informatics graduate students and summer internship program. Research areas have included consumer health informatics, medical vocabulary research and medical expert system development and evaluation.

Database Manager, University of Minnesota (1979-1980)
Modified, updated, and performed retrieval of information from the department database

Research Assistant, Biochemistry Dept, University of California, Berkeley (1973-1974)
Performed laboratory experiments to identify the essential heavy metal ion and structure of the second enzyme in the heme biosynthetic pathway

Research Assistant, Nephrology Research, VA Hospital (1972-1973)
Performed micro-experimentation on laboratory rats to measure renal function under various conditions

Teacher of English as a Second Language (1971-1972)
Provided bilingual instruction to immigrant adults in Oakland Chinatown using computer assisted instruction as an experiment to determine the usefulness of computers in teaching ESOL.

Publications and Presentations:

Divita G, Browne AC, Tse T, Cheh ML, Loane RF, Abramson M. Spelling suggestion technique for terminology servers. Proc AMIA Symp 2000; :994.

Athreya BH, Cheh ML, Kingsland LC III. Computer-assisted diagnosis of pediatric rheumatic diseases. Pediatrics 1998 Oct;102(4):E48.

Cheh ML. Internet-AI/RHEUM. A multi-media knowledge based consultation system which can be delivered over the Internet. American Academy of Dermatology Technology Conference, Bethesda, MD, November 14, 1997.

Humphreys BL, McCray AT, Cheh, ML. Evaluating the coverage of controlled health data terminologies: report on the results of the NLM/AHCPR Large Scale Vocabulary Test. J Am Med Inform Assoc. 1997 Nov-Dec;4(6):484-500.

McCray AT, Cheh ML, Bangalore AK, Rafei K, Razi AM, Divita G, Stavri, PZ. Conducting the NLM/AHCPR Large Scale Vocabulary Test: a distributed internet-based experiment. In: Masys, DR (ed). Proc Annu Symp Comput Appl Med Care; 1997 Oct 25-29, Nashville, TN. Philadelphia, PA: Hanley & Belfus, Inc. Publishers, 1987. 560-564.

McCray AT, Cheh ML. The NLM/AHCPR Large Scale Vocabulary Test. Report to the LHNCBC Board of Scientific Counselors, May 15, 1997. Bethesda, MD.

Cheh ML, Kingsland LC III, Athreya, BH. Criteria Table Expert (CTX): an environment for developing multi-media medical consultation systems. Demonstration and Invited Panel Discussion, American Academy of Pediatrics Annual Meeting, Section on Computers and Other Technologies, San Francisco, CA, October 1996.

Athreya BH, Cheh ML, Kingsland LC III. Enhancing the pediatric capability of AI/RHEUM, an expert system in rheumatology for the practitioner. American Academy of Pediatrics Annual Meeting, Section on Computers and Other Technologies, San Francisco, CA, October 1996; :9.

Cheh ML, Kingsland LC III. Criteria table representation in the medical domain. In: Kahn M, Smith J Jr, Buchanan B, Musen M, Szolovits P (eds). Working notes of the AAAI Spring Symposium Series 1992, Artificial Intelligence in Medicine. Menlo Park, CA: American Association for Artificial Intelligence, 1992; :11-24.

Kingsland LC III, Rosenberg KM, Cheh ML. CTX: the NLM criteria engine. Demonstration digest, Twelfth Annual Symposium on Computer Applications in Medical Care (SCAMC), 1988; :23-24.

Cheh ML. The expert consulting system as teacher. Invited system demonstration and discussion. Thirteenth Annual Conference of Alliance for Continuing Medical Education (ACME), New Orleans, LA, January 1988; :22.

Cheh ML. Panel on Knowledge Acquisition for Knowledge Based Systems. American Society for Information Science. Ann Arbor, MI, May 1988.

Kingsland LC III, Cheh ML, Grant KD. AI/RHEUM: four knowledge sources in a diagnostic consultant system. Demonstration Digest, Eleventh Annual Symposium on Computer Applications in Medical Care (SCAMC), 1987; :13.

APPENDIX C: LIST OF SUPPLEMENTARY MATERIAL

Genetics Home Reference Bookmark and Capabilities Brochure

Learning Aid for Exploring Genetics Home Reference

Fomous C, Mitchell J. Genetics Home Reference: Helping Patients Understand the Role of Genetics in Health and Disease. (submitted to Community Genetics and under review)

Mitchell JA, Fun J, McCray AT. Design of Genetics Home Reference: a new NLM consumer health resource. J Am Med Inform Assoc. 2004 Nov-Dec;11(6):439-47. Epub 2004 Aug 06.

Mitchell JA, McCray AT, Bodenreider O. From phenotype to genotype: issues in navigating the available information resources. Methods Inf Med. 2003;42(5):557-63.

Logan RA: Preliminary Report - Evaluation of Genetics Home Reference by Genetic Alliance. December, 2005. (not published)

Logan RA, Fun J, Cheh M. The Genetics Home Reference's image: A study of health informatics website user perceptions. (submitted to AMIA and under review)

Peng Z, Logan RA. Content quality, usability, affective evaluation, and overall satisfaction of online health information. Paper accepted by Health Communication Division, International Communication Association Annual Convention, New York, NY, May 2005.

APPENDIX D: QUESTIONS FOR THE BOARD

1. Is the current direction in development of topics appropriate? Does the Board of Scientific Counselors recommend any additional directions or advice?
2. What other promising informatics research avenues and techniques would you recommend pursuing for the development of GHR?
3. How can GHR and LHNCBC encourage collaborative informatics research on GHR content with outside groups? What communities would benefit from GHR content and how best to disseminate this information?
4. Should GHR engage in professional outreach to genetics professional organizations, such as encouraging healthcare professionals to prescribe GHR in patient/caregiver encounters in a manner similar to NLM's information RX program in which internists prescribe visits to the MedlinePlus web site?
5. Is it appropriate to focus GHR's evaluation to survey a) health information seeking consumers and b) patients/caregivers/healthcare professionals, who are motivated to obtain genetics information?