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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 LHNCBC, 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.

We would like to thank Dr. Sandra Davenport for her role in developing genetics content for the web site. Thanks also to the experts in medical genetics across the United States and Canada who provide their time and expertise to ensure the accuracy of GHR’s content.
REFERENCES


FIGURES

Figure 1: GHR Content Development Statistics

![GHR Number of Summaries on Public Site](image)

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

![Genetics Home Reference Site Traffic](image)
Figure 3: Consumer Health Information Conceptual Framework

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

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

<table>
<thead>
<tr>
<th>GHR Conditions and Subtypes</th>
<th>GHR Genes, Chromosomes</th>
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<tbody>
<tr>
<td>Achondrogenesis, type 1B</td>
<td>SLC26A2</td>
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<td>Achondrogenesis, type 2</td>
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<td>SADDAN</td>
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<td>Tetrahydrobiopterin deficiency</td>
<td>GCH1, PCBD1, PTS, QDPR</td>
</tr>
<tr>
<td>Thanatophoric dysplasia (plus two subtypes)</td>
<td>FGFR3</td>
</tr>
<tr>
<td>Trimethylaminuria</td>
<td>FMO3</td>
</tr>
<tr>
<td>Triple X syndrome</td>
<td>Chromosome X</td>
</tr>
<tr>
<td>Tuberous sclerosis</td>
<td>TSC1, TSC2</td>
</tr>
<tr>
<td>Turner syndrome</td>
<td>SHOX, Chromosome X</td>
</tr>
<tr>
<td>Usher syndrome (plus three subtypes)</td>
<td>CDH23, MASS1, MYO7A, PCDH15, USH1C, USH1G, USH2A, USH3A</td>
</tr>
<tr>
<td>Very long-chain acyl-coenzyme A dehydrogenase deficiency</td>
<td>ACADVL</td>
</tr>
<tr>
<td>Von Hippel-Lindau syndrome</td>
<td>VHL</td>
</tr>
<tr>
<td>Waardenburg syndrome</td>
<td>EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10</td>
</tr>
<tr>
<td>Weissenbacher-Zweymüller syndrome</td>
<td>COL11A2</td>
</tr>
<tr>
<td>Wilson disease</td>
<td>ATP7B</td>
</tr>
<tr>
<td>X-linked severe combined immunodeficiency</td>
<td>IL2RG</td>
</tr>
<tr>
<td>X-linked sideroblastic anemia</td>
<td>ALAS2, HFE</td>
</tr>
<tr>
<td>21-hydroxylase deficiency</td>
<td>CYP21A2</td>
</tr>
<tr>
<td>22q11.2 deletion syndrome</td>
<td>TBX1, Chromosome 22</td>
</tr>
<tr>
<td>47,XYY syndrome</td>
<td>Chromosome Y</td>
</tr>
<tr>
<td>--</td>
<td>Chromosome 1-4, 6-12, 14, 16, 17, 19, 20</td>
</tr>
<tr>
<td>Table 2: 91 MedlinePlus Topics Linked to GHR (March 31, 2005)</td>
<td></td>
</tr>
<tr>
<td>------------------------------------------------------------</td>
<td></td>
</tr>
<tr>
<td><strong>Adrenal Gland Disorders</strong></td>
<td><strong>Ehlers-Danlos Syndrome</strong></td>
</tr>
<tr>
<td><strong>Alpha-1 Antitrypsin Deficiency</strong></td>
<td><strong>Endocrine Diseases</strong></td>
</tr>
<tr>
<td><strong>Alzheimer's Caregivers</strong></td>
<td><strong>Eye Cancer</strong></td>
</tr>
<tr>
<td><strong>Alzheimer's Disease</strong></td>
<td><strong>Facial Injuries and Disorders</strong></td>
</tr>
<tr>
<td><strong>Amyotrophic Lateral Sclerosis</strong></td>
<td><strong>Fever</strong></td>
</tr>
<tr>
<td><strong>Anemia</strong></td>
<td><strong>Fragile X Syndrome</strong></td>
</tr>
<tr>
<td><strong>Arrhythmia</strong></td>
<td><strong>Gaucher's Disease</strong></td>
</tr>
<tr>
<td><strong>Ataxia Telangiectasia</strong></td>
<td><strong>Genes and Gene Therapy</strong></td>
</tr>
<tr>
<td><strong>Bile Duct Diseases</strong></td>
<td><strong>Genetic Brain Disorders</strong></td>
</tr>
<tr>
<td><strong>Birth Defects</strong></td>
<td><strong>Genetic Counseling</strong></td>
</tr>
<tr>
<td><strong>Bladder Cancer</strong></td>
<td><strong>Genetic Disorders</strong></td>
</tr>
<tr>
<td><strong>Bleeding Disorders</strong></td>
<td><strong>Genetic Testing</strong></td>
</tr>
<tr>
<td><strong>Bone Diseases</strong></td>
<td><strong>Head and Brain Malformations</strong></td>
</tr>
<tr>
<td><strong>Bone Marrow Diseases</strong></td>
<td><strong>Hearing Disorders and Deafness</strong></td>
</tr>
<tr>
<td><strong>Brain Diseases</strong></td>
<td><strong>Hearing Problems in Children</strong></td>
</tr>
<tr>
<td><strong>Breast Cancer</strong></td>
<td><strong>Hemochromatosis</strong></td>
</tr>
<tr>
<td><strong>Cancer</strong></td>
<td><strong>Hemophilia</strong></td>
</tr>
<tr>
<td><strong>Cardiomyopathy</strong></td>
<td><strong>Huntington's Disease</strong></td>
</tr>
<tr>
<td><strong>Carpal Tunnel Syndrome</strong></td>
<td><strong>Immune System and Disorders</strong></td>
</tr>
<tr>
<td><strong>Charcot-Marie-Tooth Disease</strong></td>
<td><strong>Infertility</strong></td>
</tr>
<tr>
<td><strong>Cleft Lip and Palate</strong></td>
<td><strong>Kidney Cancer</strong></td>
</tr>
<tr>
<td><strong>Colorectal Cancer</strong></td>
<td><strong>Kidney Diseases</strong></td>
</tr>
<tr>
<td><strong>Congenital Heart Disease</strong></td>
<td><strong>Klinefelter's Syndrome</strong></td>
</tr>
<tr>
<td><strong>Connective Tissue Disorders</strong></td>
<td><strong>Learning Disorders</strong></td>
</tr>
<tr>
<td><strong>Cystic Fibrosis</strong></td>
<td><strong>Leukodystrophies</strong></td>
</tr>
<tr>
<td><strong>Degenerative Nerve Diseases</strong></td>
<td><strong>Lewy Body Disease</strong></td>
</tr>
<tr>
<td><strong>Dementia</strong></td>
<td><strong>Liver Diseases</strong></td>
</tr>
<tr>
<td><strong>Developmental Disabilities</strong></td>
<td><strong>Male Breast Cancer</strong></td>
</tr>
<tr>
<td><strong>Down Syndrome</strong></td>
<td><strong>Male Genital Disorders</strong></td>
</tr>
<tr>
<td><strong>Dwarfism</strong></td>
<td><strong>Metabolic Disorders</strong></td>
</tr>
<tr>
<td>Date</td>
<td>Source</td>
</tr>
<tr>
<td>------------</td>
<td>---------------------------------------------</td>
</tr>
<tr>
<td>2/3/05</td>
<td>Course Page for BIOL 1103, Southern Wesleyan University</td>
</tr>
<tr>
<td>12/27/04</td>
<td>E-mail: physician</td>
</tr>
<tr>
<td>11/12/04</td>
<td><em>Science Magazine</em>, Netwatch column</td>
</tr>
<tr>
<td>4/6/04</td>
<td>E-mail: family member of patient</td>
</tr>
<tr>
<td>2/5/04</td>
<td>E-mail: unknown</td>
</tr>
<tr>
<td>October 2003</td>
<td>Health Sciences Library System, University of Pittsburgh Medical Center</td>
</tr>
<tr>
<td>9/10/03</td>
<td><em>Columbus Federal Voice</em></td>
</tr>
<tr>
<td>Summer 2003</td>
<td>NCI-Frederick, <em>Science Library News</em></td>
</tr>
<tr>
<td>7/8/03</td>
<td><em>The Washington Post</em>, health web site review</td>
</tr>
<tr>
<td>[ongoing]</td>
<td>Aussie Educator, Tertiary Links, Genetics</td>
</tr>
</tbody>
</table>
Table 4: Visits to GHR Pages

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

\(^1\) 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.

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

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

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

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### Table 6: Potential Outcome Variables for Future Evaluation Studies

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

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@lhc.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 Home 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 website.

   Here’s a guide to respond to all the questions in this section: In the first question, for example, if you 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)
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):
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.
Robert A. Logan

Social science analyst; Senior scholar; Director, Informatics Training

Education and Training

<table>
<thead>
<tr>
<th>Institution</th>
<th>Degree</th>
<th>Year</th>
<th>Major</th>
</tr>
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<tbody>
<tr>
<td>Tulane University</td>
<td>B.A.</td>
<td>1969</td>
<td>History</td>
</tr>
<tr>
<td>University of Missouri</td>
<td>M.A.</td>
<td>1973</td>
<td>Journalism</td>
</tr>
<tr>
<td>University of Iowa</td>
<td>Ph.D.</td>
<td>1977</td>
<td>Mass Communication</td>
</tr>
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</table>

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)


Recent Honors:


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)


Refereed Publications (since 2000):

Book Chapters

Articles and Conference Proceedings

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.

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:


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.

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.

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
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


Cardellina JH II, Famous C. Your genome—just another credit card to carry around. Fertil Steril.
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:
Inducted into the Phi Beta Kappa Society, 1999.

Publications:
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 melanogaster.

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

Publications:

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.

  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.

  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.

  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.

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:
**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 offline; 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.


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:**


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.


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.


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

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


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?