AsktheGeneticist[SM]: five years of online experience

Catherine Tesla, MS, CGC,[1] Bruce R. Korf, MD, PhD,[2] Lynn Holt, MS, CGC,[3] Sandra Prucka, MS, CGC,[2] Nathaniel H. Robin, MD,[2] Maria Descartes, MD,[2] Edward Lose, MD,[2] Adrya Stembridge, AS,[1] Michael Epstein, PhD,[1] and Stephen Warren, PhD[1]

Purpose: To identify the genetic informational needs and assess the level of awareness about clinical genetic services among adults who use the internet. **Methods:** We created an online service called AsktheGeneticist[SM] (http://www.askthegen.org) to answer questions about medical genetics. Since 2003, we have received 4497 questions from every US state and 84 countries/territories. Genetic counselors draft answers to the questions submitted. The questions and answers are next reviewed by clinical geneticists, then organized by topic and uploaded to the site. A link to an online website-user satisfaction survey is e-mailed to the user with a link to their Q&A. **Results:** Before visiting AsktheGeneticist[SM], 20% (50/247) of survey respondents were unaware that genetic services existed. After visiting our website, 23.5% (58) of survey respondents sought contact with a genetics health care professional, compared with <1% of patients who self-refer to a general genetics clinic (binomial test; $P < 0.0001$). Website users most often sought information about a known genetic condition in their family and the risk of recurrence. **Conclusions:** Our data suggest that the internet can be an effective tool for increasing the awareness of genetic services and identifying genetic informational needs of online adults, as well as for connecting patients with genetic services. ***Genet Med*** **2009:11(4): 294–304.**

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Use of the internet for medical information is rising steadily, yet little has been published regarding the use of the internet for genetics-related information (GRI).[1–4] Harris Interactive® reports the number of US adults who have ever searched for health information online has increased to 160 million. This represents 84% of all online adults.[5]

One study on the use of the internet for GRI revealed that half the patients and families surveyed searched for GRI before their genetics clinic visit, and a majority searched the World Wide Web for GRI after their visit. The number one reason cited for searching the internet for GRI was to find GRI in layperson's terms, followed by treatment information, and genetic research information. Regardless of age, race, or genetic clinic site (urban versus rural), 85% of respondents had access to the internet.[6]

The rapidly evolving nature of human genetics makes the internet a logical and effective medium for addressing questions about human genetics, not only to identify informational needs, but also to inform online users about the availability of clinical genetic services. There is growing evidence that genetic patients not only search the internet for medical information, but desire direction and input on internet use from genetic professionals. A study of patients attending a medical genetics clinic in Canada found that 80% believed that medical genetic professionals should provide them with appropriate and useful internet sites. Twenty-nine percent of patients in this study reported searching the internet for genetic information.[7] In a time of concern about the future of genetic services and the need to market the importance and availability to a broader audience, the internet presents a unique opportunity.

Advantages of the internet include convenience, unlimited access to a massive volume of inexpensive information resources, user anonymity when searching for subject-sensitive health information, ease of updating information, and the potential for interactive formats that promote understanding and retention of information.[8] This is not limited to GRI. A study examining the effect on the physician-patient relationship of public use of the internet for health information found that 85% of physicians experienced a patient bringing internet information to a visit.[9] The use of accurate relevant information was found to be beneficial and welcomed.[10–12]

Website visitors from around the world share similar goals when turning to the internet for health-related information; searches are performed for information about a specific condition, treatment, or symptoms.[13,14] When seeking health-related information, internet users most frequently cite websites from health-related government agencies, academic health science centers, and commercial health information publishers. These types of websites are viewed as offering high-quality, credible information from a trustworthy source.[8,14]

Many online genetic resources have been targeted to the medical community, the public, or both. When queried using the search term "genetic conditions," Google™ yields 50,300,000 results in 0.11 seconds (accessed, September 20, 2007).[15] For at least the past decade, there have been numerous free and fee-for-service websites that allow users to ask a doctor a question.[16] Among them are http://www.askthedoctor.com, http://www.bbc.co.uk/health/ask_the_doctor, and http://www.doctorslounge.com/ask.htm. Those websites without fees have a tendency to post previously answered questions and answers. "Ask a doctor" sites are geared toward providing general medical information and education, but are not intended to supplant the physician-patient relationship or the medical examination.

Before 2002, we were unable to locate any interactive websites geared to field questions specifically about GRI.[17] In 2003, the Department of Human Genetics at Emory University launched AsktheGeneticist[SM] (http://www.askthegen.org), a web-based resource to answer questions about genetic concepts as well as the etiology, treatment, research, testing, and predisposition to genetic disorders. In the fall of 2005, members of the Department of Genetics at the University of Alabama in Bir-

From the [1]Department of Human Genetics, Emory University, Atlanta, Georgia; and [2]Department of Genetics, University of Alabama at Birmingham, Birmingham, Alabama; [3]School of Health Professions University of Alabama at Birmingham, Birmingham, Alabama.

Catherine Tesla, Department of Human Genetics, Emory University School of Medicine, 615 Michael Street, Whitehead Biomedical Bldg, Suite 301, Atlanta, GA 30322. E-mail: ctesla@emory.edu.

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mingham joined the AsktheGeneticist[SM] team, creating a regional model for online genetics education.

SUBJECTS AND METHODS

Website overview

There are four categories of content on the website:

- Educational, including
  - previously posted questions and answers organized by topic
  - links to genetic fact sheets on carrier testing, family history, and prenatal diagnosis
  - links to other genetic resources on the web (genetic conditions, genetics education, and information for genetics professionals)
- Clinical: a guide for understanding and locating clinical genetic services
- Research: a guide for locating a genetics research study and information on what to expect when participating in a genetics research study
- Training: an overview of career options in genetics and genetics education, with links to training, certification, and professional societies

The website uses PHP (hypertext preprocessor, a reflective programming language) to generate dynamic web content, drawing question-and-answer information from an Oracle database. At present, a distinct URL (www.askthegen.org) links to both the University of Alabama at Birmingham (UAB) and Emory University existing genetic websites (http://www.genetics.uab.edu and http://www.genetics.emory. edu). Questions received on the site are e-mailed simultaneously to genetic counselors at Emory University and the UAB. Genetic counselors were trained to facilitate the operation of the website, determine which questions fit the mission of the website, and provide an internet-appropriate response to each question. Genetic counselors draft answers that are reviewed and edited by clinical geneticists, then organized by topic and uploaded to the site. Although the original goal was to present all web content at or below an eighth-grade reading level, many names of genetic conditions or enzymes involved in genetic diseases simply exceed the number of syllables allowed for this language level. For this reason, most answers are written at the ninth- to twelfth-grade level. All posted answers are reviewed and updated annually, to ensure that website content remains accurate and current.

The operation and workload of AsktheGeneticist[SM] have evolved over time. During the first 2 years the number of inquires received was small and distributed between all genetic counselors at Emory (15 at the time), essentially adding minutes of time per individual's monthly schedule. Between 10 and 30 minutes of an Emory clinical geneticist's time per week was used in the review and editing process. In the third year of the project, a grant through the Southeastern Regional Genetics Group was procured, expanding the AsktheGeneticist[SM] team to include members of the Department of Genetics at the UAB. Two genetic counselors added content and operated the website service during this time period, one from Emory and one from UAB, with the same physician oversight. As the number of questions submitted to the website increased, the combined genetic counselor time increased to an average of 4–8 hours per week. Finally, during the past 2 years of operation, one genetic counselor from Emory has been the primary genetic counselor involved with the project, with genetic counselors from UAB assisting in a lesser role. Clinical geneticists from UAB took the place of the Emory physician for the review and editing of website inquiries. At the current level of website activity (approximately 50–70 questions/week), the combined genetic counselor workload approaches 20 hours/week, with a total physician review time of 1–4 hours per week.

One of our primary concerns in the creation of this website service was human subject protection. Names and emails are not posted on the website; only the website user's question and answer. Because of the uniqueness and rarity of genetic disorders, however, anonymity may be compromised by correlation. The website disclaimer addresses this risk and includes a detailed privacy policy. There is also a warning for users younger than 18 years, since the site is not designed or intended for use by children. We have had extensive input from Emory University's general counsel and institutional review board in formulating the disclaimer.

The current form used to submit a question contains optional fields for the website user's name, city, state, country, and e-mail address. It also gives the writer the option of self-identifying as one of the following: parent of a child with a genetic disease, person with a family history of a genetic disease, carrier of a genetic disease, person affected with a genetic condition, pregnant couple, physician, student, or other. By submitting a question, the user accepts the terms of use, which include an extensive site disclaimer, copyright, and citation guidelines. These data, along with the subject of the question (e.g., inheritance, genetic testing), whether or not the question was answered (and if not, why not), and when the question was answered, are collected and stored in an Oracle database on Emory University's server. Questions we do not answer as a matter of policy include those regarding paternity, nongenetic issues, requests for medical interpretation, or those submitted by students for homework assignments. The database is accessed via an individual username and password and can be queried to obtain relevant information on items of interest, such as the number of questions from a particular geographic location or the number of questions on a particular topic.

Whether or not a question is answered, an automatic e-mail is generated based on selections from the form, then sent to the website user. The e-mail contains a direct link to the answer posted on the website, as well as a link to a website user survey. Previously posted questions and answers are searchable by category or key word from the homepage of the website.

Data collection

Data were collected on individuals who submitted a question to the AsktheGeneticist[SM] website, and individuals who visited the website without submitting a question. The Oracle database was queried to collect data on individuals who submit a question, including the volume of questions received, the number and types of questions answered, and the breakdown of all submitted questions by website user, geographic location, topic, subtopic, question concept, and whether a referral to a genetics health care provider was made.

Data on individuals who visit the website without submitting a question were captured using web analytic software. We use Google™ Analytics (http://www.google.com/analytics/) to track the ways visitors interact with AsktheGeneticist[SM]. Google™ Analytics provides in-depth website visitor behavior analysis including:

- the total number of visits and page views the site received,
- the average number of page views per visit (P/V), and the number of visits and page views over time;
- the geographic location of website users;
- the number of first-time visits and return visits; and
- the average time spent on the website.

Survey overview

We developed an online website user survey to obtain information about website usage, knowledge and satisfaction, and the demographics of genetic health seekers. The reason for collecting the survey data were to evaluate the benefit of the AsktheGeneticist^{SM} website, to determine how aware users are about the existence of clinical genetic services, and to assess the trends in the use of the internet for GRI. Before deployment, the questionnaire was reviewed by experts in statistics, public health, education, and genetics, and piloted by genetic counselors in the Department of Human Genetics at Emory University. In April 2006 a link to the survey (http://www.surveymonkey.com/second.asp?u = 800181981664) was posted on the homepage and included in the response e-mail sent to website users, along with the link to their question's answer. By completing and submitting the survey, subjects indicated their consent to participate. The study was approved by the Emory University Institutional Review Board (eIRB00003051). The survey captures a variety of types of data that can be analyzed with rather simple statistical methods, including the use of analysis of variance to understand whether the responses vary significantly across different segments of the survey population.

Potential difficulties and limitations of the proposed procedures

Web analytic data are subject to errors, since the data are based on internet protocol (IP) addresses, which are used as a surrogate for the actual human website users. Single users with dynamic IP addresses and multiple users with the same IP address are two of the ways in which IP data could be misleading.

Data collection was limited by (1) the number of unique visitors to the website; (2) the completeness of the question submission form; (3) the number of website users who choose to fill out the user survey; and (4) whether survey respondents answer all survey questions. To offset these limitations, we optimized the site to work with the most common browsers and listed the site with numerous search engines. In addition, we established reciprocal links with other genetic/medical resources on the web and published articles about the service in various local and national consumer, genetic, and other health care professionals' literature, including posters, abstracts, and platform presentations at national scientific meetings.

RESULTS

Data on questions submitted to the website

The total number of questions received from June 1, 2003 to June 1, 2008 was 4497. We received 232 questions during the first year (an average of 19 questions/month), 251 questions during the second year (an average of 21 questions/month), 827 questions during the third year (an average of 69 questions/month), 1419 questions during the fourth year (an average of 118 questions/month), and 1768 questions during the fifth year (an average of 147 questions/month). This represents a striking upward trend in the number of questions submitted over time (Wald Statistic = 14.88, $P < 0.0001$).

The overall average number of questions received per month was 75, with a high of 208 (January 2008), and a low of three (in our first month of service). Question volume peaks during February and March of each year, but the reason for this is unknown.

As of May 31, 2008, 3509 questions in 42 different categories (Table 1) have been answered by AsktheGeneticist^{SM}. We have elected to organize inquiries into broad categories (e.g., chromosome abnormalities), rather than specific, individual diseases (e.g., Down syndrome), because many questions we receive concern more than one condition.

Of the 4497 questions received, 3509 (78%) have been answered. The most common question topics are about genetic syndromes (8.6%), chromosome abnormalities (7.5%) (excluding sex chromosome anomalies, translocations, and chromosomally abnormal products of conception), neurogenetic disorders (7.3%), blood and clotting disorders (7.1%), biochemical disorders (5.8%), single gene disorders (5.2%), sex chromosome aneuploidy (4.2%), cancer genetics (4.1%), blood type (3.8%), and prenatal diagnosis and assisted reproduction (3.2%). By far the most common question is about a chromosome problem (15.4%), if we combine the related question topics of chromosome abnormalities (7.5%), chromosome abnormalities found on miscarriages (1.4%), chromosome translocations (2.3%), and sex chromosome aneuploidy (4.2%). Nearly 8% of questions received are scored as "miscellaneous." The majority of these questions are on a topic not asked about a second time. We also choose to score inappropriate, incomplete, and nongenetic issue questions in the miscellaneous category.

Questions regarding a single condition are grouped together within each category. The top condition/subject asked about within each category is also listed in Table 1.

Of the 988 (22%) questions not answered as a matter of policy, there were inappropriate questions (102), incomplete questions (95), requests for medical interpretation beyond the scope of the website (72), repeat questions (303), school projects (308), and nongenetic issues (108). Many of the questions in this category are related to nonpaternity. Those questions categorized as inappropriate include questions of a sexual, legal (i.e., I'm trying to collect child support, but I'm uncertain re: paternity), or disturbing nature. We received eight questions from an individual who stated "feeling homicidal" on four of his submitted questions. We contacted the chair of psychiatry at the university in this individual's hometown to discuss the situation. We then sent an email encouraging him to make an appointment for an evaluation.

Student submissions (31%) and repeat questions (30.7%) account for the majority of unanswered questions. This is not to imply that we have never answered any student submissions; 322 questions submitted by students have been answered. Rather, we do not answer students who seem to be posting their homework assignment to the website.

Website users who submit a question

Of the 4497 questions received, 1023 (23%) were submitted by parents of children with a genetic disorder, 758 (17%) by individuals with a family history of a disorder, 113 (2.5%) by carriers of genetic conditions, 633 (14%) by individuals affected with a genetic condition, 185 (4%) by pregnant individuals or couples, 74 (1.6%) by physicians, 83 (1.8%) by other health care providers, 765 (17%) by students, 35 (0.78%) by teachers, and 828 (18%) by individuals identifying themselves as "other." We suspect that one possible explanation for the substantial percentage of website users identifying themselves as "other" is

Table 1 Categories of questions

	Total (%)	Most common condition or subject asked about/question category
Syndromes	390 (8.6)	Waardenburg syndrome
Chromosome abnormalities	336 (7.5)	Chromosome 21
Neurogenetic disorders	329 (7.3)	Neurofibromatosis
Blood and clotting disorders	319 (7.1)	Factor V Leiden
Biochemical disorders	262 (5.8)	Galactosemia
Single gene disorders	235 (5.2)	Cystic fibrosis
Sex chromosome aneuploidy	191 (4.2)	Turner syndrome
Cancer genetics	183 (4.1)	Colorectal cancer syndromes
Blood type	174 (3.8)	Inheritance of blood type
Prenatal diagnosis and assisted reproduction	147 (3.3)	Maternal serum screening with or without nuchal translucency (NT)
Eye color and genetic diseases of the eye	145 (3.2)	Heterochromia iridium
Multiple congenital (present at birth) anomalies	137 (3)	Infants with an unclear diagnosis
Connective tissue disorders	112 (2.5)	Ehlers-Danlos syndrome
Skeletal abnormalities	108 (2.4)	Achondroplasia
Chromosome translocations	105 (2.3)	Translocation involving chromosome 21
Skin disorders	94 (2.1)	Albinism
Autism/mental retardation	93 (2.1)	Fragile X syndrome
General genetics	82 (1.8)	Questions about twins
Autoimmune disorders	68 (1.5)	Ankylosing spondylitis
Lysosomal storage diseases	65 (1.4)	Tay-Sachs disease
Chromosome abnormalities found on miscarriages	65 (1.4)	Chromosome 13
Training in genetics	60 (1.3)	What courses to take to become a geneticist
Hair	54 (1.2)	Hair color
Fetal abnormalities	34 (0.76)	Cystic hygroma
Adult-onset conditions	31 (0.7)	CADASIL
Recurrent pregnancy loss	30 (0.67)	Turner syndrome
Genetic services	28 (0.6)	How to find a geneticist
Genetic testing	28 (0.6)	How/where to get genetic testing done
Cardiovascular diseases	27 (0.6)	Wolf-Parkinson-White syndrome
Microdeletion and microduplication syndromes	23 (0.5)	Chromosome 22
Genetic genealogy	23 (0.5)	How to determine ethnic origin
Consanguinity (related by common ancestor) and degrees of relation	21 (0.47)	Cousin matings
Mitochondrial disorders	21 (0.47)	Leber hereditary optic neuropathy (LHON)
Dental abnormalities	20 (0.45)	Dentinogenesis imperfecta
Gender determination and gender identity	17 (0.38)	Androgen insensitivity syndrome (AIS)
Mental health	14 (0.3)	Schizophrenia
Ear abnormalities and hearing loss	12 (0.27)	Ear lobe inheritance
Environmental exposures during pregnancy	11 (0.24)	Diethylstilbestrol exposure
Newborn screening	6 (0.13)	General questions about NBS
Pharmacogenetics (how genes affect drug response)	4 (0.09)	Morphine resistance
Genetic/health information in foreign languages	3 (0.07)	Cancer genetic information in Spanish
Miscellaneous	353 (7.8)	Individuals with an unclear diagnosis

fear of privacy violations. Nearly 90% of website users seeking health information online are concerned that a health-related website might sell or give away information about their online activities.[16]

A study looking at patients' use of the internet before attending a medical genetics appointment found that parents referred for concerns for their child were more likely to search the internet than individuals referred for concerns regarding themselves, their pregnancy, or a more distant relative.[7] Similarly, of all questions submitted to AsktheGeneticist[SM], nearly one in four came from parents of children with a genetic disorder.

Issues genetic health seekers ask about

Most individuals seeking GRI had a tendency to ask about a specific concept. Among these concepts were the following: explanation of a genetic condition, which includes questions about the natural history and prognosis of a disorder (872, 19%); risk of recurrence (631, 14%); information regarding a possible diagnosis including questions requesting a diagnosis through the website based on information provided by the website user (533, 12%); inheritance of a disorder (292, 6.5%); testing for a disease, including questions about what genetic testing is and how and where to obtain genetic testing (289, 6%); medical management of a genetic condition (251, 5.6%); questions regarding laboratory test results (227, 5%); whether two or more health issues in the same individual are related to one another versus coincidental (165, 3.7%); available treatment for a genetic condition (142, 3%); where to find more information, including questions on website resources, support groups, treatment centers, and how to locate other patients with the same diagnosis (114, 2.5%); the prognosis for a specific genetic condition (99, 2.2%); whether conditions in a family are related to one another versus coincidental (63, 1.4%); prenatal diagnosis (55, 1%); assisted reproduction and/or the availability of preimplantation genetic diagnosis (41, 0.9%); research studies (29, 0.6%); consultation on a case involving a patient (6, 0.1%); and other (688, 15%).

In this study, one in three genetic health seekers are looking for an explanation of a genetic disorder and/or the risk for recurrence of a genetic disorder. Many times this information is readily available on websites such as the Genetics Home Reference website, GeneReviews, or MedlinePlus®. When the information is already available, we provide links to any such reliable existing information on the internet, along with our brief answer.

Many genetic health seekers who submit questions to AsktheGeneticist[SM] could benefit from the services offered by a genetics health care professional, such as genetic counseling or a genetic evaluation to rule a specific condition in or out. AsktheGeneticist[SM] is neither designed nor put forward as being a substitute for an in-person visit with a genetics specialist; rather, we intend to complement or supplement the information received from a genetics health care provider. In response to the 4497 questions received on AsktheGeneticist[SM], we recommended that 1711 (38%) of the website users have genetic counseling or a genetic evaluation with a clinical geneticist. To that end we provide them with links and instructions for the National Society of Genetic Counselors, GeneClinics, and American College of Medical Genetics websites. For those genetic health seekers residing outside the United States, we suggest an appropriate website link or phone number of the nearest genetic center, when available.

Examples of submitted questions

The following is an example of a question about a known diagnosis in a family, with our answer directing the website user to a genetics health care professional:

Q: For many years now, my family has suffered with retardation problems. In a family of five children, three are retarded, one suffers from bi-polar problems and then there is me. After the marriage of one of my retarded siblings and the subsequent birth of his two children, genetic testing was done on my mother and two of my siblings and a genetic deformity was found that was associated with height and mental capabilities. After a recent batch of testing, another genetic deformity has been noted in my brother's family (wife and 2 children) that my mother refers to as KGB or KBG. Is there some place that I can look these deformities up on the internet? I am concerned that the deformity may be dormant in my children or grandchildren but may surface in later generations.

A: From what you are describing, we are assuming that some members of your family have been diagnosed with KBG syndrome. KBG syndrome is a collection of findings including shorter than average height, moderate to severe mental retardation, skeletal and dental problems, and characteristic facial features.

KBG syndrome is inherited in an autosomal dominant fashion. This means that only one copy of a nonworking gene is needed to have the syndrome. When a parent has KBG syndrome, there is a 50/50 chance with each pregnancy to have a child with KBG syndrome. For unknown reasons, men have been reported with this condition more than women.

Some persons with KBG syndrome have an involved and severe presentation, whereas others are mild with few health problems. This range of expression can make it confusing when one is trying to determine whether they may have signs of KBG syndrome or not. For this reason, we would encourage you to consider having an evaluation with a clinical geneticist to be sure that you do not have minor features of KBG syndrome. This would then answer your question as to whether your children and grandchildren are at risk.

There is a website that lists genetic clinics by geographic location: www.geneclinics.org; click on "clinic directory." Information about genetic counseling and how to find a genetic counselor in your area are available online from the National Society of Genetic Counselors at www.nsgc.org. Click on "consumer information" or "find a counselor." You can also search for genetic health care providers on the American College of Medical Genetics website at: www.acmg.net; click on "find a geneticist" and then enter your geographic information.

This next example illustrates how AsktheGeneticist[SM] does not provide specific medical advice, but rather gives relevant information along with a recommendation for referral:

Q: I am a male, 36 years old, whose mother passed away from extremely aggressive inflammatory breast cancer. I have been reading that I should be tested for certain cancer genes including those for breast cancer and colon cancer. Is that something that is recommended? FYI there is also a family hx of a maternal grandfather with skin cancer (but didn't die from it), another grandmother died of lung cancer, and I have previously had a basal cell carcinoma removed.

A: Meeting with a cancer genetic counselor for risk assessment is the best way to determine whether you are a candidate for cancer susceptibility gene testing. We will provide some basic information here, as well as how to contact a cancer genetic specialist in your area.

First, cancer is a common disease, and all of us have about a 30% (1 in 3) chance of developing any type of cancer over the course of our lives. Women have a one in 8 (12%) chance of developing breast cancer during their lifetime. Male breast cancer makes up <1% of all cases of breast cancer. The majority of male breast cancer cases are diagnosed between 60 and 70 years of age. One in 17 persons will develop colorectal cancer sometime during their life.

Most cancers occur as sporadic conditions (i.e., occur by chance) and are not hereditary. For example, approximately 90–95% of the cases of breast cancer is sporadic and thought to be due in part to the cumulative effects of multiple factors such as lifestyle and environmental exposures. In contrast to sporadic cases, 5–10% of breast and ovarian cancer cases are due to an inherited susceptibility.

Characteristics of hereditary cancer include an earlier age of onset (usually under age 50), several affected family members in two or more generations on the same side of the family, multiple primary cancers in the same person, bilateral cancer (cancer in paired organs like both breasts) and rare cancers in the family (male breast cancer). You are not automatically at an increased risk for cancer susceptibility; more information about your family is needed for an accurate assessment. We recommend for you to have cancer genetic counseling. Please visit the Cancer Genetics Service Directory to locate a provider in your area at http://www.cancer.gov/search/geneticsservices/.

There is a website that lists genetic clinics by geographic location: www.geneclinics.org; click on "clinic directory." Information about genetic counseling and how to find a genetic counselor in your area are available online from the National Society of Genetic Counselors at www.nsgc.org. Click on "consumer information" or "find a counselor." You can also search for genetic health care providers on the American College of Medical Genetics website at: www.acmg.net; click on "find a geneticist" and then enter your geographic information.

A final example of a question submitted illustrates that some visitors to AsktheGeneticist[SM] have already seen a genetics professional but are asking for clarification after their visit:

Q: We have been told that our daughter "most likely" has a mild form of Cardio Facio Cutaneous syndrome. The geneticist said that a research facility looking at her DNA has found a gene sequence anomaly nearby the problem genes already attributed in confirmed cases to CFC syndrome. This together with her appearance and developmental delay and the fact that they were looking in this area for a problem (and found one) leads them to suggest that the diagnosis is most likely outcome. How likely? Can you help clarify?

A: The geneticist you are working with is the best person to address your questions. We will provide some basic information here. Cardiofaciocutaneous (CFC) syndrome is a condition that involves many organ systems and is characterized by heart abnormalities, characteristic craniofacial findings, and skin abnormalities. Other health problems and developmental delay are also seen. There is wide variability in the clinical presentation of individuals with this condition. The diagnosis of CFC syndrome is determined based on clinical findings and genetic testing. The four genes known to be associated with CFC syndrome are *BRAF* (~75–80%), *MAP2K1*, and *MAP2K2* (~10–15%), and *KRAS* (<5%). It is unclear at this time if all the genes associated with CFC syndrome have been identified. This, combined with the variability in clinical presentation, may help you understand why you are being told your daughter most likely has a mild form of CFC syndrome.

Location of AsktheGeneticist[SM] website users who submit a question

The 4497 questions received on AsktheGeneticist[SM] originate from all 50 of the United States plus the District of Columbia (Total = 3553; Table 2), and 84 countries/territories (Total = 944; Table 3). There were 369 website users who specified the US as their country of residence, but did not specify their city or state.

The percentage of questions received from outside the United States (19%) has remained relatively steady since the inception of our service. Within the United States, Californians submitted the most questions (460), followed by residents of Georgia (425), Texas (310), Florida (307), New York (234), and Ohio (190). Questions originating outside the United States came largely from website users in the United Kingdom (333), Canada (261), Australia (185), India (181), and the Philippines (34). There were 14 website users who did not specify a location (city, state, or country).

Website traffic (from Google™ Analytics; includes all website users)

During the most recent year (June 1, 2007 through May 31, 2008) for which data are available, there were 82,140 website visits. A visit is defined as a series of actions that begins when the visitor views the first page from the server, and ends when the visitor leaves the site or remains idle beyond a preset time limit (i.e., 30 min). For the same period, there were 374,429 page views, or a total of 4.56 page views/visit. A page view is defined as the number of times a page was viewed by visitors. The average amount of time genetic health seekers spent on the website was 3 minutes, 44 seconds.

The majority of website users during this time period found the site using an unpaid search engine (72.19%). Others found AsktheGeneticist[SM] while visiting another website (9.49%), and 18.33% came to the site by typing the URL directly into their web browser. The majority of website visitors are new (88.42%), whereas 11.58% are returning visitors.

The geographic location of website users tracked by the web analytic software over the past year mirrors the location of website users who submit a question. Approximately 77% (63,151) of website visitors are from the United States, with the highest number of visitors residing in California (6339), New York (5135), Texas (4454), Georgia (3674), and Florida (3216). Website visitors outside of the United States (18,989 or 23%) originated from 165 countries/territories, with the highest number from the United Kingdom (4474), Canada (3880), Australia (1744), India (1028), and the Philippines (997). This is a much broader reach than the 84 countries/territories from which questions originated.

Website user survey

As of May 31, 2008, there were 247 complete survey responses. Seventeen individuals began but did not complete the survey. For this reason, their responses were not tabulated in our data. Our results reveal that most survey respondents found the website either via a search engine (67%) or a link from another site (25.5%), have visited the site once (79%), submitted a question to the website (90%), read previously posted questions and answers (77%), are female (80%), between the ages of 35 and 44 (24%), white (78%), and reside within the United States (72%). This profile of an AsktheGeneticist[SM] website user is consistent with the typical online health information seeker.[16,18,19]

Table 2 Number of questions received within the United States by state

AK	11
AL	80
AR	52
AZ	82
CA	460
CO	71
CT	55
DC	13
DE	16
FL	307
GA	425
HI	7
IA	49
ID	39
IL	176
IN	93
KS	31
KY	51
LA	71
MA	144
MD	86
ME	26
MI	169
MN	58
MO	91
MS	26
MT	13
NC	180
ND	8
NE	38
NH	27
NM	19
NJ	174
NV	34
NY	234
OH	190
OK	30
OR	72
PA	169
RI	15
SC	76
SD	16
TN	100
TX	310
UT	25
VA	118
VT	12
WA	93
WI	73
WV	35
WY	6
?	369

Other survey respondents indicated that they are American Indian or Alaskan Native (5.3%), Asian (9.7%), Black or African American (4%), Native Hawaiian or Pacific Islander (0.4%), or Other (10.5%). Eighteen (7.3%) of the survey respondents indicated that they are Hispanic or Latino. We have received one question in Spanish and one question in French since inception of the website. In these two instances the questions were translated and responses were posted back to these individuals in their native language. All other submitted questions have been in English.

Forty-seven percent (117) of survey respondents indicated that they visited the website to find out information about a known genetic condition in their family, and 46% (114) indicated that they visited the website to find out information about a possible diagnosis for themselves, their child, or other family member. Other reasons for visiting AsktheGeneticist[SM] included searching for more information about genetic testing or screening (23.5%), collecting information for a research or school project (14.6%), or coming across the site while surfing the internet (10%). A small percentage of website users were referred to the website by a friend or associate (2%).

Survey respondents most often viewed the question and answer pages (82%), followed by Links to Genetic Resources (47%), Genetic Fact Sheets (44%), Genetic Research Studies (27%), Clinical Genetic Information (11.3%), Careers in Genetics (6.5%), or None of the Above (12.5%). For those sections of the website visited, most users rated the information as extremely helpful or helpful. Nearly 75% are very likely or likely to recommend the website to someone else.

A total of 78 survey respondents gave specific suggestions to improve the site: 22 requested that any/all questions be answered, since theirs was not; 32 indicated that the website was "very helpful," "quick and to the point," "valuable," "informative," "easy-to-understand," "great," "fabulous" and did not have any suggestions for improvement; 11 made comments about the turnaround time for answering questions (too long); one was a request to post information on avian influenza; one was a request for PowerPoint presentations about medical genetics; one was a request to increase the size of the text box for submitting a question; three were requests to send them periodic updates to their e-mail addresses on topics they are interested in; two comments related to the "search" feature of the website,

Table 3 Number of questions received outside the United States by country

Amer Samoa	2
Argentina	2
Austria	1
Australia	185
Azerbaijan	2
Bahrain	1
Bangladesh	2
Belgium	4
Belize	2
Bosnia	2
Brazil	20
Canada	261
China	8
Columbia	1
Congo	2
Costa Rica	2
Croatia	3
Cyprus	2
Czech Rep	1
Dominica	2
Ecuador	3
Egypt	11
Eritrea	1
European Union	3
France	8
Gambia	4
Germany	3
Ghana	3
Greece	21
Guatemala	2
Holland	1
Hungary	6
India	181
Indonesia	5
Iran	13
Iraq	6
Ireland	31
Israel	15
Italy	8
Jamaica	1
Japan	2
Jordan	10
Kenya	1
Korea	2
Kuwait	7
Lao's People Democratic Republic	2
Lebanon	2
Macedonia	1
Malta	1
Malaysia	9
Mauritius	1
Mexico	8
Namibia	1
Netherlands	5
New Zealand	22
Nigeria	14
Norway	4
Pakistan	24
Philippines	34
Poland	2
Portugal	5
Puerto Rico	9
Qatar	1
Romania	8
Russian Fed	3
Saint Lucia	2
Saudi Arabia	13
Serbia	1
Singapore	6
Slovenia	2
South Africa	13
Spain	6
Sri Lanka	2
Sudan	1
Sweden	12
Syria	4
Taiwan	3
Thailand	3
Trinidad	2
Tunisia	2
Turkey	4
UK	333
Wallis and Futuna Islands	1
Zaire	3
Zimbabwe	1
?	16

with a preference for the use of "and" rather than "and/or" terms; one comment stated that the "wording in the answers is sometimes too technical and long"; one comment mentioned that "information and answers are vague, and should be more in depth"; and three comments were unintelligible.

Before visiting AsktheGeneticist[SM], 20% of survey respondents were not aware of the existence of genetic services (i.e., physician geneticists, genetic counselors). As a result of visiting the website, 23.5% (58) of survey respondents sought contact with a genetics health care professional, compared with <1% of patients who self-refer to a general genetics clinic (personal communication, Emory University Genetic Clinics) (binomial test; $P < 0.0001$).

DISCUSSION

Millions of people turn to the internet for health information—more internet users have sought medical information on the web than have shopped online, looked up stock quotes, or checked sports scores (Fox, 2002). Cross-culturally, studies indicate that the majority of online health seekers look for information on a particular condition because either they or someone they know has been diagnosed with the condition.[8,9,19] In our survey, 47% (117/247) of respondents indicated that they visited the website to find information about a known genetic condition in their family, and 46% (114/247) stated that they visited the website to find information about a possible diagnosis for themselves, their child, or other family member.

Although many online health seekers view the internet as an important information source, physicians remain the primary source of medical information for almost all patients.[19] The body of answered questions at AsktheGeneticist[SM] amounts to a library of information about various aspects of medical genetics and specific genetic conditions intended to complement, but not replace, the information received from genetic and other health care providers. Table 1 shows that certain categories (e.g., chromosome abnormalities) received more questions than others. This may reflect a lack of available online/offline information on certain topics, a difficulty understanding certain concepts or mechanisms of particular types of genetic disorders, a lack of prognostic information for a disorder, or an explanation from a health care provider that needed more emphasis or reiteration for one or more of these reasons.

Earlier in this report, we described the advantages of using the internet for communicating genetics information. In our experience, there are also some disadvantages to using the internet to answer questions about genetics. These include reliance on the website user to provide accurate medical information and history, lack of knowledge about the website users' education level, lack of knowledge about the website user's intentions for obtaining and using the information we provide, lack of knowledge as to whether the information will be shared with their health care provider, and not knowing if the person writing in is who they say they are.

Concern about unequal access to the internet is one of the major often-cited complaints about internet-based education. Some would say that this is not an issue, since public libraries and schools are well equipped with internet access in low-income areas, even when there are few other resources available to people in these locations. The term "digital divide" was coined in the 1990s to describe gaps in access and understanding that excluded certain groups (for socioeconomic or geographic reasons) from the use of digital technologies such as the internet. Our survey data comes from respondents in all age groups, including: 15 and under (4.8%), 16–24 (8.5%), 25–34 (19.8%), 35–44 (21%), 45–54 (21%), 55–64 (15.4%), 65–74 (3.6%), and 75 and over (0.81%). Less than 4% of all AsktheGeneticist[SM] website visitors (Google[TM] Analytics) connect to the internet using dial-up, whereas the majority connect via cable modem (58%), DSL (24%), or T1 line (12%). Those who responded to the website user survey had a DSL connection to the internet (32.8%), followed by cable modem (31.3%), and a T1 line or better (5.7%).

Most website user survey respondents are white (78%) women (80%). This is not surprising as women have been found to search the internet for health information more frequently than other groups, as reported in a 2008 study conducted by comScore, Inc., a global internet information provider.[20] Other studies agree that women tend to be more proactive than their male counterparts when searching for health care solutions, as women make over 75% of the health care decisions for the family.[21,22] In a study examining the relationship of race to women's use of health information resources, white women were more likely than black women to use the internet, print news media, and health policy organizations.[22]

Some questions submitted to AsktheGeneticist[SM] indicate that the website user has already seen a genetics health care provider, but the user still has questions or needs clarification on a topic such as inheritance. The nature of many of these inquiries would not necessarily require another visit with a genetics health care provider. Furthermore, it may prove troublesome for a patient to contact a genetics health care provider with a question about a nonemergency issue after weeks, months, or even years have passed since the initial visit. An online interface with a genetics health care professional may prove more appealing than the circular nature of phone menus and layers of administrative personnel patients often wade through to obtain the information they seek. The convenience, cost-effectiveness, and continuity offered by the internet are reflected in the fact that a growing number of medical insurers are now willing to cover online consultations.[23]

The data from our website user survey suggest that the internet may be an effective tool to both inform website users of the existence of clinical genetic services and also to connect individuals and families with such services in their geographic region. An unexpected finding has been that 20% (50/247) of survey respondents were unaware of the existence of genetic services before visiting AsktheGeneticist[SM].

Genetics on demand

When compared with other medical specialties, there is a dearth of "genetics on demand," i.e., access to GRI on the web, in the form of online genetic communities, news feeds, blogs, and video libraries. In a list of the 20 largest health websites ranked by a combination of Inbound Links, Google Page Rank, Alexa Rank, and US traffic data from Compete and Quantcast, genetic websites are not among them.[24]

Within the past year, however, an online Community of Genetic Educators has been launched by the National Human Genome Research Institute. This online community is a "place for genetic educators to meet, collaborate, share and learn."[25] During its first 6 months, this website has attracted 325 members (accessed June 30, 2008). In addition, the Genetic Alliance provides both a discussion list for patients and their families to access online meeting spaces for various conditions, as well as a Network Forum, which connects organizations within the Genetic Alliance.[26]

The genetics-specific news feeds we located did not come from genetic organizations, but instead were available from general medical or technology information websites, such as

Medical News Today,[27] *Technology Review*,[28] and *Nature News*.[29] Corresponding blogs about genetics are also available from these websites.

As more people have access to high-speed internet connections, there has been a surge in the use of video to deliver information, including medical information. For example, a set of videos depicting the early signs of autism is available on the Autism Speaks (www.autismspeaks.com) website, and the Harvard Medical School Center for Neurofibromatosis and Allied Disorders has a video of a consultation with a clinical geneticist for neurofibromatosis (http://www.understandingnf1.org/). A search on YouTube™ for videos on genetic conditions yielded videos on a number of genetic disorders, including phenylketonuria, hemochromatosis, fragile X syndrome, neurofibromatosis, Huntington disease, epidermolysis bullosa, alpha-1-antitrypsinase deficiency, cri-du-chat syndrome, Duchenne muscular dystrophy, and cystic fibrosis (accessed May 13, 2008).[30] The quality and accuracy of information in these videos certainly varies; however, the potential for widespread dissemination of information should not be underestimated. Globally, YouTube™ ranks third (Alexa Rank, accessed May 13, 2008)[31] among all websites, behind Yahoo! and Google™.

Genetics has a major role to play in all aspects of medicine and health, yet our study demonstrates a lack of knowledge of the existence of genetic services in 20% of website survey respondents. As mentioned earlier, there is a concern about the future role of genetics in medicine. More than one study has identified a shortage of genetics-trained health professionals and the need for an "ultimate makeover" to integrate genetics into medical practice.[32,33] Marketing is an educational process. AsktheGeneticist℠ illustrates a novel way to educate and thereby market the importance and availability of genetic services around the globe to prospective patients, other medical professionals and individuals interested in a career in genetics. Based on our online experience with AsktheGeneticist℠, we offer three recommendations. Since the internet is widely used to search for GRI, we suggest directing patients and professionals to reliable websites by providing a list of recommended websites including AsktheGeneticist℠ as part of your clinical practice, and including the URLs as links on your institution's website. More research is needed to compare the efficacy of an online question-and-answer format versus other modes of access to genetics health care providers (e.g., online educational videos, telephone consultation, telemedicine, in-person genetic counseling). Even so, we recommend considering online access to genetic professionals as part of your clinical practice. This may not only reveal the amount of information patients retain after a genetic consultation, but may also uncover areas in need of more emphasis or reiteration. Finally, the availability of clinical genetic services and how to locate them should be widely publicized, so that individuals, families, and other health care providers know how to access them.

Future direction

Answering questions from around the world via AsktheGeneticist℠ has provided a glimpse into the types of genetic information people are seeking, who they are, and where they reside. There are still many unknowns. For example, what types of problems do people face with regards to obtaining desired information on genetic disorders? How do website users access genetic services after receiving an answer from AsktheGeneticist℠? What is their perception of the quality of health information from an in-person consult when compared with the internet or other sources (e.g., radio, television)? What other websites have they looked for information? How many people discuss the information gained from AsktheGeneticist℠ with their family and/or health care providers? Moving forward we plan to expand our online website-user survey to elicit responses to these and other relevant questions. Without a doubt the internet is changing how people access genetics information. Our hope is that "genetics on demand" will continue to increase along with our understanding of the best ways to communicate with and better understand the genetic informational needs of website users.

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