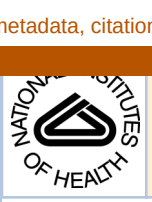

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How Parents Search, Interpret, and Evaluate Genetic Information Obtained from the Internet

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Abstract

This study describes how parents of a child referred for genetic services searched the Internet for information, summarizes how they interpreted and evaluated the information they obtained, and identifies barriers that they encountered. Audio-taped interviews were conducted with 100 ethnically diverse families referred to a pediatric genetics clinic. After transcription, coded text was entered into a software program (QSR N6) for searching and data retrieval. Matrices were created to systematically categorize and compare families' Internet use. Eighty-three percent of families obtained Internet information about the diagnosis, the clinic visit, and/or treatment and services. Those not conducting searches lacked access, Internet experience, or a diagnostic term and had lower incomes and less education, regardless of ethnicity. Families sought information in preparation for the clinic visit but barriers to obtaining and interpreting relevant information were common. Parents' Internet searching experiences illustrate common barriers to obtaining and understanding genetic information. Identifying them can help genetic counselors facilitate parents' searches for relevant information.

Keywords

Genetic counseling; Genetic services; Health knowledge; Attitudes; Parents; Practice; Internet; Consumer health information; Health literacy; Qualitative research

Introduction

Parents whose child has a disability or chronic health problem search for information about diagnoses, prognoses, treatments, and supportive resources from a variety of sources (Bailey and Powell 2005; Pain 1999; Stark and Moller 2002). This information helps parents adjust to an uncertain future (Hummelinck and Pollock 2006; Lewis et al. 1991), fulfill a perceived responsibility to become the "expert" on their child's condition (Skinner and Schaffer 2006), and cope with the emotional impact of the diagnosis (Perrin et al. 2000; Schwartz et al. 2006; Skirton 2006; Taanila et al. 1998). The search for information varies with parents' educational level, their previous experience with and knowledge of the condition, their relationship with their child's physician, the severity of their child's disease and the length

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of time since a diagnosis was made (Hummelinck and Pollock 2006; Noll et al. 2001; Stark and Moller 2002; Taanila et al. 1998).

Parents' searches for information have become increasingly influenced by the Internet. In 2006, an estimated 113 million American adults searched the Internet for health information (Madden and Fox 2006). Estimates for parents' e-health usage (i.e., using the Internet and related technologies to search for information related to health and health care services) range between 20% and 65% (Ikemba et al. 2002; Rahi et al. 2003; Tuffrey and Finlay 2002; Wainstein et al. 2006).

Having a diagnostic term to search for health information has been associated with Internet use (Christian et al. 2001; Schaffer et al. 2008; Tuffrey and Finlay 2002), while lack of a diagnostic label, low medical and genetic literacy, and limited awareness of appropriate sites hinder attempts to find relevant information (Aslam et al. 2005; Christian et al. 2001; Dhillon et al. 2003; Schaffer et al. 2008). Although racial/ethnic inequities in Internet access exist, their impact on computer access and searching has been relatively small as compared to that of higher education level, prior Internet experience, and younger age. (Bansil et al. 2006; Diaz et al. 2002; Murray et al. 2003; Rice 2006).

The Internet has dramatically increased parents' access to what was once privileged health information, potentially changing their expectations and affecting their relationships with health care professionals (Berland et al. 2007; Murray et al. 2003; Sillence et al. 2007; Skinner and Schaffer 2006;). Relatively few studies have documented the methods parents use to search the Internet, the types of information they find, whether it meets their expectations, and the impact on health care decisions. Although the potential for harm worries some professionals (Hart et al. 2004), families view the Internet as increasing their ability to communicate with physicians and improving their understanding of medical conditions (Bansil et al. 2006; Murray et al. 2003). In general, e-health users evaluate the Internet as a valuable source of information. For example, 84% of parents who used the Internet prior to a pediatric outpatient visit evaluated the information they obtained as useful (Tuffrey and Finlay 2002). Even parents who had low expectations of obtaining relevant results evaluated their searching experiences as positive and rated the information they obtained as "easy to find" and "helpful", regardless of its relevance (Aslam et al. 2005;Dhillon et al. 2003; Murray et al. 2003; Taylor et al. 2001). In a survey of over 3,200 respondents, Murray et al. (2003) found that while some users remained concerned about the accuracy of the information and were unsure how to interpret the information they found, only 1% described Internet searching as harmful.

The degree to which patients apply the information they obtain from the Internet is uncertain. In one survey, up to 20% of e-health users agreed that the information they obtained had an impact on their health care decisions (Madden and Fox 2006). Others have reported that parental access to information, by itself, did not necessarily alter health care decisions nor did it automatically improve their understanding of their child's condition (Wainstein et al. 2006). When practitioners assessed parents' knowledge about their child's surgery, increased understanding was associated with higher education levels and the seriousness of the surgery, but not with access to Internet information (Noll et al. 2001).

Clinical experience suggests that many parents of a child referred for a genetic evaluation and counseling visit search the Internet for information before coming to clinic. A survey of a general genetics clinic population found that 47% searched hoping to find information in layman's terms, but 41% who searched characterized the information they obtained as confusing and difficult to understand (Taylor et al. 2001).

This report summarizes how parents searched the Internet for information about their child's condition before and after their referral to a university-based pediatric genetics clinic. It describes searching techniques and targets, the impact of the information obtained, and the barriers parents faced in finding relevant information. Recognizing the barriers that families face to accessing and understanding genetic information can help genetic professionals identify and address their clients' informational needs.

Methods

Recruitment

The data reported here on Internet use come from a larger, longitudinal, ethnographic study that examined how parents who had a child or who were at risk for having a child with a genetic disorder sought out, understood, and used information over time to interpret genetic disorders and to make decisions about reproduction, health care, and services. Between 2000 and 2002, families were recruited from those newly referred by their health care providers or developmental evaluation centers to a pediatric genetic clinic located in a university-based hospital in the south-eastern United States. Our aim was to obtain a sample of families who represented the wide variety of indications typically seen in an active pediatric genetics clinic in the U.S.; thus, we recruited families irrespective of diagnosis. Families were purposively recruited to represent the ethnic and socioeconomic diversity of the clinic population. After the initial clinic referral, the families were sent a recruitment brochure by the first author (MIR). Those interested in the research project returned the detachable postcard indicating their willingness for a research assistant to contact them with further information about the study. Approximately 30% of those recruited were enrolled. After they had the opportunity to discuss the study with a member of the research team, parents were sent a copy of the consent form. Both Spanish and English versions of the brochure and consent form were sent to families who were Latino. An ethnographer reviewed the consent form with the family in person either before their first clinic visit or, when the initial clinic session was not observed, before beginning the first interview. Interviews with Latino families were conducted in either Spanish or English, as they preferred. We invited mothers, fathers, and other interested family members to participate in the interviews. Except for one grandmother, the biological mother was the primary respondent in the families. This was due, in part, to the fact that mothers tended to be the primary care givers, health care managers, and e-health users in their families. Participants were compensated $50 for each observed clinic visit and interview. This study was approved by the appropriate Institutional Review Board and informed consent was obtained from all participants.

Procedures

The study began with families' first visit to the clinic. Because the larger study was interested in families' understandings and evaluations of information conveyed in the clinic visit, ethnographers, primarily graduate students and the project PI (DS), observed and wrote extensive field notes on the communications and interactions between families and the clinic staff (including genetic counselors, medical geneticists, and trainees) during their first and any subsequent counseling and evaluation session. This observation was followed by an in-depth, semi-structured interview conducted approximately 4 weeks after the initial visit, and three subsequent interviews at approximately 6 month intervals. The initial interview questions asked families to evaluate the clinic session and to describe where they had searched for information, services, and supports in relation to the child's condition; their understandings of the condition and its ramifications; and decisions made related to health, reproduction, and services. Since the Internet was frequently mentioned in conjunction with these topics, we continued to ask about Internet use in the subsequent interviews that were designed to elicit information about changes in information seeking and understanding over

time. Interviews typically lasted between 60–90min and were conducted in the home or other place of the family's choosing. Audio recordings of the interviews were transcribed verbatim and Spanish interviews were translated into English.

Data Analysis

Using a grounded theory approach, the authors and two research assistants conducted a preliminary analysis of the interview data to find out where families searched for information relevant to their child's condition. In this analysis, the Internet emerged as a major resource in parents' quests for a diagnosis, prognostic information, treatments, services, and supports. Using QSR N6, a software program designed for qualitative data analysis, we coded and collated all information related to the Internet for each family. We then examined the collated interview data for a closer, more systematic analysis of parents' Internet use. To do this, we independently read all relevant data and created data display matrices (Miles and Huberman 1994) that allowed us to systematically categorize how parents' searched the type of information sought, and their views of the advantages and barriers to Internet use. If there was no reference of Internet use for a family, we reviewed the full set of transcripts to confirm that no Internet searching had been done. Employing the constant comparative method (Goetz and LeCompte 1981; Patton 2002) and negative case analysis (Denzin 1989), we used the matrices to compare families case by case and created accounts of parents' patterns and evaluations of Internet use.

Results

Participants

Information about Internet use related to their child's condition was obtained from 100 families out of a total of 105 families enrolled in the study. Although a few families were bi- or tri-racial, the primary respondent, most often the mother, identified herself as being African American (20), Latino (11), Native American (7), and White (62) (see Table 1 for demographic information). Families represented a diverse range of income and educational levels. Nearly half (45%) had annual incomes of less than $30,000, while only a quarter (26%) had incomes of $60,000 or greater. Six percent of the mothers had less than a high school education, 26% had a high school diploma or equivalency, 33% had some college or an associate's degree, 24% had a bachelor's degree, and 11% had advanced degrees. Thus, almost two-thirds of the mothers had not completed a college degree.

The interviews conducted with the mother or other primary caretaker was the focus of the analysis since 96% of the probands were children ranging in age from 1 month to 18 years. Sixty-five percent of the children were 5 years of age or younger. Fifty-four of the probands had a diagnosis that was confirmed by genetic testing or had phenotypic features highly suggestive of a specific clinical diagnosis while 42% had a non-specific condition for which a genetic etiology was possible but could not be verified. The remaining 4% had conditions that were ruled out by genetic testing. Table 2 provides examples of the probands' diagnoses.

Prevalence of Internet Use

The vast majority of the families, 83%, obtained information from the Internet about their child's diagnosis or symptoms, the clinic visit, and/or available treatment and services. While 69% of the respondents searched the Internet themselves, an additional 14% received Internet information from family, friends, co-workers, doctors or other service providers. Internet use varied by ethnicity, income, and education (see Table 3). Fewer African American (65%) and Latino (55%) families reported searching the Internet themselves as compared to Native American (86%) and White (94%) families. Those with lower incomes

and less education, regardless of ethnicity, were less likely to have ever used the Internet to search for information. Ninety-four percent of college-educated families searched the Internet themselves as compared to only 44% of those with a high school education. Eighty-nine percent of families with household incomes greater than $30,000 a year searched the Internet themselves as compared to 44% of those with earnings below that amount. However, another 24% of low-income families received Internet information from others.

The 31% of families who did not conduct an Internet search themselves said that they lacked access, were not confident in their computer searching skills, had no diagnostic term to use, were not concerned about their child's condition, had little confidence that they would find relevant information, or were unaware that the Internet could provide medical or genetic information.

How Families Searched the Internet

Searching was widespread but varied in efficiency and effectiveness. Parents, typically the mother, began the search by using one or more of the child's symptoms or when available, diagnostic terms, as key words to plug into widely available search engines such as Google or Yahoo. Other websites sponsored by large health or advocacy groups (AMA, Web MD, NORD) were also used in searches. As one mother explained: "What I do is, I basically get on the Internet and whatever my son's symptoms are that seem unusual to me, I put it in and see what it pulls up." Some parents searched by using specific diagnostic terms such as "fragile X," "iso chromosome," or "partial 4p monosomy" while others used general terms such as "genetics" or "mental retardation". These latter searches retrieved large amounts of information that required interpretation in order to judge their relevance—a task that was difficult for parents to accomplish on their own. One mother summarized her experience by saying the Internet has "a lot of information with no good way to filter or decipher how you fit [it together]."

While prior Internet experience and familiarity with medical and genetic terms were important factors, the availability of a specific diagnostic term to use in searches played a key role in parents' ability to find information. By comparison, parents who lacked a specific diagnostic term or who had been given a term that led to few relevant links were forced to conduct searches based upon non-specific symptoms, if they searched at all. The lack of a readily available "name" to use in searches limited the information parents obtained. One mother explained: "Not having a diagnosis has made it very hard for us… because we have nothing to put into the computer."

Upon learning the name of a suspected or confirmed diagnosis, some families immediately turned to the Internet. One mother of a child with Prader–Willi syndrome said: "Five minutes after we got the diagnosis… we got on the computer. I didn't know what else to do." For families that fit this pattern of searching, their initial success at finding information led to subsequent searches during which they looked for new information about management or treatments. Other events that triggered additional searching included learning of a new diagnostic term, the appearance of new symptoms, or an upcoming clinic visit.

Information learned during the clinic visit became a source of new terms that parents then used in searches. In some cases, this new term allowed them to find more specific information about diagnostic criteria, available treatments and management, genetic testing and potential results, inheritance patterns, and potential risks for relatives. But even when their child had a confirmed diagnosis, the term did not always lead to a successful search if it was not descriptive enough to link to relevant information or if it was difficult for parents to remember or spell. Some diagnostic terms led directly to websites that provided

information for a specific genetic condition (e.g., Prader–Willi syndrome, NF-1) while others did not. For example, a mother of a child with 45,X/46,XY mosaicism did not remember the name of the diagnosis. She did recall the phenotypic description provided by her local doctor that her son "might have both sex organs," but admitted "I didn't really know how to look that up." Medical terms that described portions of the child's phenotype (e.g., aniridia, Hirschsprung disease, agenesis of the corpus collosum) became key words that enabled parents whose child did not have a specific diagnosis to find some information. As parents gained more experience in searching for information, some improved their searching skills by using more effective key words and by becoming better judges of the relevance of the information they retrieved. One parent summarized this learning curve: (Now) "I put in 'partial trisomy 8p'. I found out that if I put in just 'chromosome 8', I don't necessarily get anything."

Some families in the study had been given a tentative diagnosis and came to clinic for confirmation. But after the visit, parents who continued to believe that their child's features matched a particular diagnosis searched for information about the condition even when the appropriate genetic tests failed to confirm it. Other families stopped searching after they received genetic test results that did not support the tentative diagnosis as they found the test results sufficiently reassuring to convince them that their child did not have the suspected condition. Parents described ending a search after they encountered disturbing photos, especially those involving physical malformations, and frightening descriptions of the suspected diagnosis. When confronted with these negative images, some parents concluded that their mildly affected child could not possibly have the same diagnosis and they quit searching. A mother of a child suspected to have Klippel–Trenauny–Weber syndrome fit this pattern. After seeing photos of affected children on the Internet, she said: "This is not what we have…pictures of these grotesque looking port wine stains, you could tell it was so much different than what (my son) had… I didn't like what I read, so I stopped." When their child's diagnosis had been confirmed in the distant past, parents' goals in searching focused on finding prognostic information and services that would promote their child's health and development. Some families stopped using the Internet to search for medical information, but continued to use it to connect to advocacy agencies and parent support groups. Parents also described reaching a point where continued searching seemed futile because they were unable to find answers to their remaining questions. One mother of a child with a chromosome abnormality explained:

> (The Internet) doesn't really have what I'm looking for. I know what'sit's like to be the parent of (an affected) child. I need to know (how to) help them succeed, help them be productive citizens. This is going on, have you seen this, what can I do? And I haven't found that and I probably haven't looked enough. That may be out there, and it may be at some of these websites. I just have not taken the time to hunt it down.

Although some families stopped searching, others continued even in the face of little apparent progress, fearing that if they stopped searching they might miss a vital piece of information. One mother whose child had unexplained developmental delay said, "As a mom, I would want more information. I want everything explained, every little detail, as much information as I can possibly get."

Overall, while parents demonstrated varying degrees of success at finding answers to their questions, they remained grateful for the opportunity that the Internet provided to allow them to access information independently of their child's health care provider. Most parents remained optimistic that the answers to their questions existed but eluded them because of their inadequate searching skills. Parents who found a site they trusted and that provided

information they could understand repeatedly returned to it rather than seeking out new sites.

Which Information Was Sought?

The targets of parents' searches are summarized in Table 4. Few parents searched for information specifically about the genetic aspects of a condition and their potential reproductive risks. This finding may have been influenced by the high proportion (42%) of the probands who had no specific diagnosis and by the distribution of inheritance patterns in those with a confirmed or highly suspected diagnosis. In the latter category, most (75%) were caused by new mutations with low recurrence risks for parents. Seven percent of the diagnosed disorders were inherited in an autosomal recessive pattern but the parents of these affected children did not plan to have more children. Only 6% of the cases were clearly the result of autosomal dominant or X-linked recessive inheritance with the remaining cases having more than one possible mode of inheritance.

Preparing for the genetics clinic visit was a target of some searches. These parents described the searches as allowing them to learn a "genetic vocabulary" so that they would be better prepared to understand the discussion during the visit and enabling them to ask appropriate questions. A mother of a child with osteogenesis imperfecta explained:

> We had no control over what was happening to her and from a parent's standpoint that's a very frightening experience…the more you understand it, the more you have the ability to deal with it and overcome it. We wanted to be able to understand what the doctors were telling us…what is involved, what can we expect…how can I ask the right questions?

A mother of a child with velo-cardio-facial syndrome described how she took Internet information to the geneticist, saying to him: "These are my questions. I found that on the Internet…from NORD… explain this to me." Unlike this mother, however, only a few families told the genetic clinic staff about their Internet searching. The clinicians themselves rarely initiated a discussion of this topic with a family nor did they fully explore the family's searching experience when they were told about it. But in the interviews, parents described how they had found scientific and/or medical terms during their searches that they tried to use during the clinic visit so that the geneticist and other medical professionals would take them more seriously. A mother of a child with an undiagnosed condition explained:

> If you talk to doctors as if you have just an ounce of education, they're gonna talk to you like you have an ounce of education, and they're not gonna BS you…. When I talk to doctors I sound totally different than how I talk everyday. I don't want them to think I'm some country bumpkin that doesn't know my thumb from my elbow. I mean, I've done a lot of research and I want them to know it. I don't want them to repeat the same information to me that I already know.

Other families found the Internet to be of limited help in preparing them for the clinic visit because of the over-whelming amount of information they retrieved. A mother of a child with unexplained developmental delay said: "I went on the Internet … (but) it was too much information to try to figure exactly what they were going to do. We really didn't know what to expect." Even those families who had found information about a specific diagnosis before the visit reported feeling blindsided when the health professional confirmed the diagnosis during the visit. The advanced preparation helped them to understand the information presented but this only partially prepared them for the emotional impact of hearing the diagnosis confirmed. Still, they judged themselves to be better off than parents who had not prepared at all. A mother of a child with a chromosome deletion explained:

> I was at a definite advantage…being able to go to the Internet, which has been a godsend, (to find out): "Here's what a translocation is; what (it) looks like; what an unbalanced translocation looks like; why it is not good to have an unbalanced translocation." I went in speaking the same language that the counselor was speaking. For anyone (without) access to the Internet, it would definitely be very intimidating.

Advantages of and Barriers to Internet Searching

Parents cited convenience, privacy, and the ability to find previously unobtainable information as the main advantages of using the Internet. One mother of a son who has a clinical diagnosis of autism noted:

> I can't wait to get (on the Internet) and be able to find out things….the convenience of it being in my house… I can set (my son) on the bed without having to think, "Well, I've got to load him up and go to the library. I won't go today, because I won't have the time". So I'm looking at a new world.

Parents who had conducted successful searches evaluated the Internet information they obtained as being superior and more current than that available from printed sources, especially when the condition was rare. One mother of a child with Turner syndrome said: "The (sites) that I did finally find were really helpful… as far as up-to-date information … stuff that I don't think I'd have found elsewhere."

Parents also readily acknowledged multiple barriers to using the Internet to search for the kinds of information that they desired. They expressed frustration at determining which term to use as a key word in a search engine because general terms retrieved too much information yet specific terms could yield nothing of relevance or no information at all. Families beginning a new search typically under-estimated the amount of genetic and medical information they would retrieve using general search engines and were surprised at how much of it was either not relevant or contained specialized words and concepts that they could not interpret. A mother of a child with a 22q deletion said: "The only thing that I know is that chromosome 22 is the one that they've mapped. I went to that web page because I thought 'won't it be interesting'? (But) I had no idea what they were talking about."

Even when parents had been given a specific diagnostic term, they had trouble remembering it and spelling it. One mother said that when her pediatrician first mentioned neurofibromatosis, "I had no idea how to spell it. I looked it up in this book (and) took that spelling so I could find out how to put it on the Internet. That's how I got to the website for the NF Foundation." During their searches, parents discovered that a condition, such as hearing loss, could have different etiologies, several different terms could refer to the same phenotype (e.g., VCF, 22q deletion, DiGeorge, or Shprintzen syndrome), and a diagnosis could have such broad phenotypic ranges that only very limited natural history information was available (e.g., many sub-microscopic chromosome abnormalities).

But parents who learned or were able to overcome these barriers could benefit from the Internet's capability of providing access to multiple sources of information that allowed them to become experts about their child's rare disease. They used this information to educate others, including their child's doctor, about the disorder and its prognosis and management. One mother described this experience:

> I've done most of my research on the computer because it was such an unknown condition. Nobody knew what 'chiari' was. I didn't have anybody locally that could tell me. Doctors didn't know what it was. There's two forms of chiari. One has to do with the intestinal and digestive tract. The other one has to do with the

neurological system. One is pronounced ‘chiari’ and one is pronounced ‘kiari’, but they’re spelled the same. When I asked [my doctor] about chiari, he started spouting off about the digestive condition and I said, no that’s not it, you don’t know what I’m asking you about, so just forget. So, I actually knew more than the doctors did by simply looking it up on the Internet and it’s been wonderful.

Discussion

The emergence of the Internet as a tool for obtaining health information has had a profound impact on the amount and kinds of information that parents who have a child with a (suspected) genetic condition can now obtain. In this study, parents made extensive use of the Internet to find information. Over two-thirds (69%) of the families searched the Internet themselves for information about their child’s condition while an additional 14% received Internet information from others. This rate of Internet use is somewhat higher than the estimates reported for a variety of medical specialties (Ikemba et al. 2002; Noll et al. 2001; Rahi et al. 2003; Tuffrey and Finlay 2002;Wainstein et al. 2006), including two previous studies of a medical genetics clinic (Christian et al. 2001; Taylor et al. 2001). The respondents in the current study were parents, mostly mothers, the majority of whose children had a specific diagnosis—all three characteristics have been associated with higher Internet use (Bansil et al. 2006; Christian et al. 2001; Madden and Fox 2006; Rice 2006; Taylor et al. 2001). This high rate of use, found among parents who were representative of a diverse, university-based pediatric genetics clinic population, especially with respect to education and income, is significant.

Parents’ Internet searches were triggered by events such as an upcoming clinic visit, a new diagnostic term, and the emergence of a new symptom. Parents cited both the convenience and the widened access to previously unobtainable information as critical features in their decision to use the Internet as a searching tool. Factors associated with Internet searching included higher educational levels, familiarity with Internet searching for other kinds of information, the presence of a diagnostic term, the degree to which they were able to obtain relevant information, and the time since diagnosis. Consistent with previous studies, our results suggest that ethnicity alone was not an important factor in Internet searching as compared to educational level. Few families cited their inability to access the Internet as an obstacle.

The parents in this study represented a heterogeneous population who differed in many factors that influenced how or when a family used the Internet and determined whether or not they were actively searching for information at the time they were interviewed. When they spoke about their decision to stop searching, parents either stated that they had found what they were searching for or they described recurring barriers to finding relevant, understandable, or new information. Two commonly described barriers that families faced in this study were the need for sophisticated searching methods in order to find relevant information and the difficulty in interpreting the complex information once it was obtained. As other studies have reported (Schwartz et al. 2006; Sillence et al. 2007; Wainstein et al. 2006), key word searches using common search engines retrieved an overwhelming amount of information, much of which parents found un-interpretable and irrelevant. Some parents improved their ability to find information and judge its relevance over time, but many continued to have difficulties with these tasks. When their child had a confirmed diagnosis of a relatively common genetic condition, parents were more successful at finding some relevant information, especially if there was at least one website dedicated to it but locating information about the less common conditions remained difficult.

The findings from this study suggest that parents who search for information about their child's potential or confirmed genetic diagnosis have multiple informational needs. First, as Holloway et al. (1997) have also noted, families would benefit from easily accessible and comprehensible information about what to expect during a clinical genetics visit, including the types of questions that will be asked of them and a list of potential questions that other families in similar circumstances have found useful to ask. Second, while both patients and practitioners have a limited knowledge of genetic principles, (Brunger et al. 2000; Chapple et al. 1995; Giardiello et al. 1997;), there are many sources that attempt to provide this information. But families and their referring physicians are also unfamiliar with the goals and tools of clinical genetics (Roche 2006). Providing this information before a visit could facilitate the genetic counseling process *irrespective* of whether or not a specific diagnosis had been made. Explaining critical concepts such as variable expression and genetic heterogeneity in layman's terms could strengthen families' understanding of testing options and potential results (see Table 4). If this background information were readily available and written in the context of a clinical genetics visit, and in a patient-friendly way, genetic counselors and geneticists could then dedicate more time during the visit to the specifics of the particular case. Third, a list of commonly used genetic and medical terms including correct spellings and their meanings could enhance parents' ability to conduct more effective searches prior to the visit. This successful advanced preparation could make the clinic visit more effective and increase families' satisfaction. Families would also benefit from guidance on how to assess a website's credentials and from an explanation of how biased ascertainment can influence the information's validity. Equally important, methods of disseminating new and existing information about genetic testing and screening should be tested and evaluated in order to capitalize on the ways that families typically search for information. Novel ways to deliver genetic information, for example, the use of Wikipedia and other commonly used Internet resources, should be considered to broaden its accessibility.

The results of this study suggest that when families had the opportunity to prepare for a genetics clinic visit, they perceived they had a better understanding of the genetic and medical information that was communicated during the session. Most families cannot fully absorb information, either intellectually or emotionally, while it is presented during a clinical encounter (Collins et al. 2001; Strauss et al. 1995). And while access to appropriate information does not, by itself, necessarily improve understanding or change health care decisions (Noll et al. 2001;Wainstein et al. 2006), it has been reported to help parents emotionally adjust to their child's disability (Taanila et al. 1998). Our findings suggest that computer-based information, if more widely and thoughtfully used, could better prepare families for their genetic evaluation and counseling visit by reinforcing, supplementing, and improving their understanding of the information they obtain both inside and outside the clinic. Several features of computer-delivered information are well-suited to parents' information needs, especially at the time of an initial diagnosis: around-the-clock accessibility, user control of the amount and type of information delivered, and the endless capacity for repetition. Although computer-based information cannot replace a knowledgeable provider (Green et al. 2001; Westman et al. 2000), not all families have access to providers who are familiar with their child's genetic condition. At a minimum, parents and their providers could benefit from a highly accessible source of balanced information that addresses misconceptions about genetic conditions (e.g., "genetic disorders cannot be treated") and the goals of a clinical genetic evaluation. These misconceptions prevent practitioners from referring appropriate families for genetic evaluation and counseling (Sikkens et al. 2002) and for education and therapeutic services (Schaffer et al. 2008).

Providing clinically relevant and easily accessible information to families before and after the clinic visit can significantly enhance the goals of genetic counseling by increasing parents' ability to understand the information presented during the clinic visit and facilitating their ability to review it following the clinical encounter. Although some authors have suggested that providers direct families to appropriate websites (Diaz et al. 2002; Murray etal. 2003; Schwartz et al. 2006; Taylor et al. 2001; Wainstein et al. 2006), this recommendation may not be feasible for some genetic conditions and websites vary greatly in their ability to communicate information to less educated users. The formidable barriers facing parents, especially those with lower educational and income levels, and their local providers to finding and interpreting relevant information for uncommon conditions, remain challenging. As for the large number of families whose child remains undiagnosed, information that described the implications of negative genetic testing results, the value of re-contacting the genetics center in the future, and specific strategies to locate resources to support their child, could be useful.

While some inaccurate and threatening information is available on the Internet and remains a concern, genetic clinicians should view the Internet as an opportunity to extend their reach outside of the clinic. During the clinic visit, genetic professionals should routinely inquire about Internet information that the family has obtained so that misinterpretations can be corrected and accurate information can be reinforced. Many parents have concerns that health professionals may disapprove of their searching (Hart et al. 2004; Murray et al. 2003). By initiating an open discussion, clinicians can validate the parents' roles in seeking information. This discussion provides an opportunity to guide families to better sources, for example, by providing relevant key words and accurate spellings. Referring providers would also benefit from this guidance. Specifically discussing parents' Internet searching experiences can also enlighten the clinician as to the kinds of information that parents are currently accessing and define the kinds of information families would like to find. This process can help identify the health literacy barriers the family may face. Finally, genetic centers should consider collaborative efforts to pilot, test, and evaluate examples of the patient information they provide in order to judge its educational effectiveness.

Although the majority of participants in this study were parents, their Internet searching experiences are likely to be representative of other patients referred for genetic services. One limitation of this study is its timing as recruitment ended in 2002 and Internet use, in general, has continued to grow during the intervening time period, making it highly likely that the usage rates reported in this study are now underestimates. The number and quality of websites dedicated to specific genetic conditions and the number that provide basic genetic information have also increased since the time of this study making it possible that more families are finding the information they seek. But while knowledge of genetic principles may have continued to improve in the general population, it is likely that low genetic literacy remains a substantial barrier to finding and interpreting information.

Internet access and use continues to expand in parallel with the amount and complexity of information parents need to understand their child's genetic condition. These trends will only increase the difficulties that families face in retrieving relevant and accurate information. Barriers that prevent families' understandings of medical and genetic information threaten their ability to improve their child's quality of life. Genetic professionals should take a leading role in identifying and addressing these barriers and help families enhance their abilities to find the information they seek.

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Table 1

Demographics (*N*=100)

	Percent
Mother's ethnicity	
African American (20)	20
European American (62)	62
Latino (11)	11
Native American (7)	7
Mother's education level	
<9–12 years (6)	6
HS Grad/GED (26)	26
Some college (22)	33
College grad (24)	24
Graduate school (11)	11
Household income	
<15,000 (26)	26
15000–29,999 (19)	19
30,000–59,999 (29)	29
60,000–89,999 (10)	10
90,000 and over (16)	16
Age of probands	
0–5 years (65)	65
5–18 years (31)	31
18 or over (4)	4

Table 2

Probands' Diagnoses and Reasons for Referral

Chromosome abnormalities
45,X
47,XXY
47, XX, +21
mos 45, X/47,XYY/46,XY
mos 47,XX, +13/46,XX
Partial trisomy 8p
Partial monosomy 18q; partial trisomy 4q (*de novo*)
Partial monosomy 9q;partial trisomy 19p (pat)
Del 4q (*de novo*)
Del 15q (Prader–Willi Syndrome) (*de novo*)
Del 22q11 (VCFS/DiGeorge Syndrome) (*de novo*)
Del 22q13 (*de novo*)
Clinical phenotypes
Aniridia
Arrhinea, microophthalmia
Blepharophimosis
Cleft lip and palate[a]
Congenital cataracts[a]
Fetal alcohol syndrome/Fetal alcohol effects
Hallux valgus[a]
Hirschsprung disease
Klippel–Trenauny–Weber
Lissencephaly
MD, unknown type[a]
Microtia
Robin sequence
VATER
Single gene disorders
Cherubism
Cystic Fibrosis[b]
Ectodermal dysplasia
Ehlers-Danlos syndrome[a]
Freidrich ataxia R/O[a]
Hemophilia A R/O[a,b]
Hereditary Hearing Loss (Connexin 26)[b]
Huntington Disease R/O[a,b]

	Hypochondroplasia[a]
	Hypophosphatasia[a]
	MODED
	Multiple epiphyseal dysplasia[a]
	Neurofibromatosis type 1
	Neurofibromatosis type I R/O
	Osteogenesis imperfecta
	Torsion dystonia[a,b]
	Tuberous sclerosis[a]
Non-specific diagnoses	
	ADHD
	Ambiguous genitalia
	Autism
	Asperger
	Developmental delay
	Gastroschisis
	Hearing loss
	Hypotonia
	Immune deficiency/lymphangiectasia
	Infantile spasms
	Mental retardation/abdominal wall defect
	Speech delay

R/O ruled out by testing or clinical exam

[a] Positive family history

[b] Confirmed by genetic testing

Table 3

Internet Use: Ethnicity, Household Income and Mother's Education Level

	Searched themselves	Had others search	Total obtaining Internet information
Ethnicity			
African-American (20)	11 (55%)	2 (10%)	13 (65%)
European American (62)	51 (82%)	7 (11%)	58 (94%)
Latino (11)	4 (36%)	2 (18%)	6 (55%)
Native American (7)	3 (43%)	3 (43%)	6 (86%)
Household income			
$0–$29,999 (45)	20 (44%)	11 (24%)	31 (69%)
$30,000–$59,000 (29)	23 (79%)	3 (10%)	26 (90%)
$60,000–$89,000 (10)	10 (100%)	0 (0%)	10 (100%)
$90,000+(16)	16 (100%)	0 (0%)	16 (100%)
Mother's Ed. Level			
<9th–12th grade (7)	2 (29%)	1 (14%)	3 (43%)
HS grad/GED (25)	13 (52%)	6 (24%)	19 (76%)
Some col./AA degree (33)	22 (67%)	5 (15%)	27 (81%)
College grad (24)	21 (88%)	2 (8%)	23 (96%)
Adv degree (11)	11 (100%)	0 (0%)	11 (100%)

Table 4

Targets of Parents Searches

Prepare for the genetic clinic visit
Find a description of process
Identify which questions they would be asked
Research which questions they should ask
Identify which professionals would be involved
Learn about genetic testing options
Identify the tests that might be done and who should be tested
Learn about how the sample is obtained
Find out about the accuracy and validity of the test
Learn about the potential results from particular tests
Learn about the implications of test results for relatives
Learn about diagnostic and prognostic information
Find information about physical, medical, and behavioral characteristics of a diagnosis
Learn about the prevalence, natural history, lifespan, quality of life, and etiology of a diagnosis
Learn about differential diagnoses and the range and variability of symptoms
Assess the likelihood of particular diagnoses
Learn about support services
Locate other families, especially those with older children
Identify and obtain appropriate services
Read about other families' experiences
Learn about what insurance is likely to cover or deny
Learn about management and treatment
Compare the availability and effectiveness of various treatment and therapies
Locate experienced centers/providers
Find clinical trials
Read about research advances