

# Parents Go Online to Understand Genetic Diagnoses

The Human Genome Project has led to discoveries of the genetic basis for hundreds of disorders. It also has advanced the understanding of basic genetic processes at the molecular level. At the same time, the Internet offers broad access to a wide range of genetic and medical information. How do parents who have young children with suspected or known genetic disorders seek out and use this knowledge? The Culture and Family Interpretations of Genetic Disorder Project—an ethnographic study of 106 families referred to a pediatric genetic counseling and evaluation clinic—sheds light on this question and on implications for early interventionists. This *Snapshot* summarizes the study's findings, which were reported by Dr. Debra Skinner and Rebecca Schaffer of FPG in a recent issue of *Infants & Young Children*.



## Study Participants

- Families were recruited from a pediatric genetic clinic in the southeastern United States. They represented the ethnicity and socioeconomic diversity of the clinic population.
- Of the 106 mothers or other primary informants in the families, 62 were European American; 23, African American; 13, Latino; and 8, Native American.
- Forty-seven percent had annual incomes of less than \$30,000, while 21% had annual incomes of more than \$75,000.
- Seven percent of the mothers did not have a high school diploma; 26% had a high school diploma or GED; 34% had some college or an associate's degree; 23% had a bachelor's degree; and 10% had advanced degrees.
- Sixty-five percent of the children referred to the clinic were 5 years of age or younger, with the average age of this group being 30 months.
- Forty-four families did not receive a clear diagnosis of a genetic condition, whereas 62 families received or confirmed a definitive genetic diagnosis through genetic testing or a clinical examination.

## Families' Internet Use

- Eighty-three families used the Internet for some purpose related to their child's condition.
- Sixty-nine families went online themselves.
- Fourteen families had others search the Internet on their behalf.
- Ninety-four percent of European American families used health resources on the Internet, followed by 86% of Native American families, 65% of African American families, and 55% of Latino families.
- Of families with annual household incomes below \$30,000, 69% reported using the Internet.
- Among most families, the mother was the primary or sole user of online health resources.

## Why Families Go Online

Families used the Internet for reasons that varied over time. Before obtaining a diagnosis for their child's condition, some mothers conducted searches of conditions and characteristics to diagnose the disorder or to identify medical specialists who could. Some mothers also said they relied on the Internet to gain scientific literacy, which they used during clinic visits so that professionals would treat them as knowledgeable clients. A few parents who felt overwhelmed or uncomfortable asking questions in the clinic turned to the Internet for explanations in layman's terms, prognoses, or case descriptions posted online by other families. The majority of the parents did not turn to the Internet until they had a potential or confirmed diagnosis, which provided a term to plug into a search engine. The Internet also served families beyond the diagnosis, helping them identify and access services and treatments.

Although most parents with a confirmed diagnosis used the Internet with less frequency and intensity once they had a solid understanding of the diagnosis and treatment options, about 10% remained avid users. They became active in online support and advocacy groups. These avid users were either families of children with extremely rare genetic conditions or families who searched the Internet for alternative opinions on treatment plans.

Parents of children with rare disorders used the Internet to find other families whose children had the same diagnosis. Noting that their clinicians had difficulties offering prognoses, they turned to other families in hopes of finding similar cases that might offer more information.

If parents did not agree with medical geneticists about treatment, they sought second opinions from researchers, clinicians, and other families on the

Internet. They also found research needed to negotiate with their geneticists and other healthcare providers about treatment options.

For most parents, the Internet added to what they learned from healthcare providers. But it functioned as primary information and support for parents who said that they did not receive sufficient information about their children's diagnosis and treatment options or that their concerns had not been acknowledged by doctors.

## Effects of Internet Use

Going online directed parents to networks of other parents who provided the experiential knowledge they found most useful in making decisions about their child's care. Parents also increased their own scientific literacy through Internet research. These parents saw themselves as information managers who weighed Internet content not only on scientific merit but also as to how it meshed with their firsthand knowledge of their child's condition. They came to view professionals as only one source of information rather than the seat of authoritative knowledge.

Some parents shared information they had downloaded with their doctors or service providers to educate them about their child's condition. Parents also took printouts to their doctors to obtain help assessing the information's accuracy and relevance, although it was more common for parents to turn to other parents in online communities associated with specific disorders to evaluate the quality of information and to determine what practices worked optimally with children.

Because of the rapid growth of genetic knowledge and its availability on the Internet, genetic diagnosis has assumed more value. With a diagnosis, parents have a term to plug into the Internet, which in

turn guides them to the basic genetics of the condition, potential treatments or interventions, and access to virtual research, advocacy, and support. Families commonly reported using a diagnosis to research a condition online and then talking with teachers and other service providers so that they could better understand how to work with the child.

Although parents said they benefited from online research, they still experienced anxiety when online resources presented contradictory advice, did not answer questions, drew a negative portrait of their child's future, or kept them searching for a missing piece of information. They felt a keen responsibility to find information that would make a difference in their child's health and development.

## Implications

In terms of how genetic discoveries will influence early intervention, scholars and practitioners have discussed how early interventionists will need to assist families in accessing and making sense of genetic information related to a disorder and tailoring interventions to the specific genetic disorder. Practitioners will need more information about basic genetics, genetic counseling, and supports and therapies for particular disorders, as families will be searching for this kind of information. This study shows, however, that this knowledge no longer belongs solely to the professional. These parents may know more than most professionals about their child's specific genetic disorder. They may have spent far more time doing the research and talking with other parents about optimal treatments and interventions, and come to see their role to be that of educating service providers about what they have learned. In effect, they are "citizen scientists" who share authoritative knowledge with service providers and expect to be partners in decisions affecting their children. ■

## To Learn More

This snapshot is based on:

Skinner, D. & Schaffer, R. (2006). Families and genetic diagnoses in the genomic and Internet age. *Infants & Young Children, 19*(1), 16-24.