Some people doubted President John F. Kennedy when he told the United States that man would land on the moon before “the sun set on the sixties.” Similarly, some people questioned the goal of completing the sequence of the human genome by 2003. The Human Genome Project (HGP), an international research effort to complete the sequence of human DNA (Collins et al., 1998), saw public- and private-sector researchers competing to map the human genome and ultimately finish ahead of schedule. The successes of the HGP have enabled researchers to pinpoint errors in genes, the smallest units of heredity, that cause or contribute to disease. We now know that heritable variations in genes contribute not only to rare conditions but also to a host of common conditions such as heart disease, diabetes, Alzheimer’s disease, and many types of cancer and mental illnesses (Collins, 1999). In other words, it is now believed that all diseases have a genetic component, whether inherited completely (e.g., cystic fibrosis) or resulting from the body’s response to environmental stresses such as viruses or toxins, or a combination of both (e.g., most cancer cases; Lou, 2002).

What does the advance in genetics mean to the occupational therapy profession? Clinically, as scientists gain more and more genetic information and a better understanding of gene–environment interactions, occupational therapy practitioners can play a role in preparing individuals to adjust to probable life changes and make needed lifestyle modifications. Just like the Americans With Disabilities Act (1990), the Individuals With Disabilities Education Improvement Act (2004), and Medicare, the fruits of the HGP will provide opportunities for occupational therapy practitioners to engage in new and exciting areas of clinical practice and research.

In the 2007 Eleanor Clarke Slagle Lecture, Jim Hinojosa voiced that in this era of “hyperchange,” the profession of occupational therapy “cannot afford to ‘wait and see’ . . . that we must both plan ahead and think fast” (pp. 629–630). The profession to date has made some effort to assist practitioners in applying genetic information and promoting genetics literacy, but we do not possess a clear plan detailing how occupational therapy will integrate and use the massive amount of genetic information being published, nor do we appear to be thinking quickly enough about how occupational therapists can arm themselves with the clinical and research skills necessary to
become part of the genetic expanse. The rapidity at which the science of genetics is progressing is astounding, and the time is now to ask what occupational therapy will look like in the age of the human genome. The issues are: (1) How can the profession equip its clinicians and researchers with basic language and tools to ethically use genetically based information, and (2) how do occupational therapists take advantage of opportunities to participate in genetic research studies? The option to ignore the genomic expanse is clearly not viable. The purpose of this article is to discuss some specific areas of genetic research that have the most obvious opportunity for occupational therapy research and practice and to suggest mechanisms of capitalizing on these opportunities.

Psychiatric Genetics

Psychiatric genetics is a specialized area of genetics that focuses on the inherited factors involved in psychiatric and behavioral disorders. Four paradigms are currently conceptualized in the area of psychogenetic research: basic epidemiology, applied epidemiology, molecular genetics, and gene finding.

The fields of both basic and applied genetic epidemiology are concerned with quantifying the degree of trait heritability and exploring the nature and mode of action of genetic risk factors (Kendler, 2005). Research in these fields is dominated by twin and adoption studies that focus on understanding the relationship between genes and the environment and how this relationship changes over time. The twin study design specifically allows for the teasing apart of genetic and environmental influences by mathematically calculating differences between monozygotic twin pairs (who share 100% of their genetic code) and dizygotic twin pairs (who share an estimated 50% of their genetic code). Evidence for environmental influences is supported when monozygotic twins are dissimilar for a specific disease or psychopathology.

For example, Gatz et al. (2006) evaluated the genetic and environmental influences on Alzheimer’s disease in a sample of 392 twin pairs in which one or both twins had Alzheimer’s disease. Results confirmed previous findings for high heritability of the disorder but also found that nongenetic risk factors played an important role in the risk and onset of the disease. The authors concluded that environmental factors should be the focus for interventions to reduce risk or delay the onset of disease. This study, however, did not go so far as to indicate which environmental factors might best influence or predict Alzheimer’s disease. It may be important to know whether factors such as frequent engagement in active exercise or individual sleep patterns are important to consider in distinguishing at-risk people who do and do not develop Alzheimer’s disease. The twin modeling design would allow researchers to ask those questions and to explore how lifestyle and environmental experiences influence the risk of developing psychiatric disorders and functional limitations because of those disorders. Here is a role designed specifically for occupational therapists and one that should be of great interest to the profession. Occupational therapists are trained not only to understand the mechanisms of the brain and body but also to recognize how the disease process can both influence and be influenced by the occupations or activities in which one engages. This knowledge and training uniquely positions occupational therapists to contribute to psychiatric genetic research: The ability to explore environmental influences on the development of disease and on the impact of function should appeal to many occupational therapy researchers.

Recent advances in psychiatric genetics have also supported sensory modulation disorders as distinct and heritable conditions. Goldsmith, Van Hulle, Arneson, Schreiber, and Gernsbacher (2006) found that both auditory and tactile defensive ness were modestly heritable, with both genetic (38% and 52%, respectively) and shared environmental (33% and 17%, respectively) influences. In addition, these researchers found that although these forms of sensory defensiveness were moderately genetically associated with fearful temperament and anxiety, they were relatively genetically distinct from other common dimensions of childhood behavioral dysfunction. This finding suggests that sensory defensiveness may be an independent diagnostic condition (or set of symptoms) not accounted for in the current psychiatric Diagnostic and Statistical Manual of Mental Disorders (American Psychiatric Association, 2000). This finding should be of great interest to researchers investigating the origins of SPDs and to occupational therapy practitioners working with children with sensory defensiveness. This information not only enhances our understanding of SPDs but may also serve as valuable information for therapists to challenge insurance companies who deny payment for treatment of these disorders. Collection of twin data over time will potentially shed light on how SPDs change over time and the effects of gene–environment interactions in the progression of these disorders. Occupational therapists have the potential for contributing at the level of psychiatric genetics research, specifically by asking and answering questions related to functional outcomes, predicting environmental factors that may lead to differences in those outcomes, and contributing knowledge related to clinical phenotypes (manifestation or presentation of the disorder) and brain–behavior patterns.

The two other categories of psychiatric genetics are molecular genetics and gene finding. Molecular genetics is concerned with the identification of DNA variants (variation between two chromosomes) and tracing pathways from DNA to disorder. Gene finding is less biological and more statistical and is focused on determining the location and identity of susceptibility genes (Kendler, 2005). The work in these two areas spans psychiatric genetics and general genomic research and is discussed in relation to the HGP.

The Human Genome Project

The purpose of the HGP, which was officially completed in 2003, was to identify all of the approximately 20,000 to 25,000 genes in human DNA and determine the sequences of the 3 billion chemical base pairs that make up human DNA. By making this map and sequence available to the public domain, the HGP has helped to accelerate the identification of genes involved in a variety of medical and psychiatric conditions (Collins & McKusick, 2001). Predictive genetic testing allows the
how participation in meaningful occupations contributes to health and well-being. These programs could then be evaluated for their effectiveness to determine whether such preventative methods can decrease health complications for a majority of at-risk patients. If this is the case, such research could provide evidence that occupational therapy services are also cost-effective.

Ethical, Legal, and Social Issues

New genetic knowledge raises concern about patients’ rights, privacy, psychological well-being, and the philosophical nature of knowing what the future of what one’s own health holds (Lapham, Kozma, & Weiss, 1996; Rothstein, 1998). The increased availability of genetic testing will affect not only the people who choose to take the test but also those who cannot afford it, do not want it, or do not understand it. What does it mean to a person to be told that she has a gene that increases risk for stroke, heart disease, or Alzheimer’s disease? Should she tell her family, especially her children? If so, at what age and what does she tell her children? How does one tell spouses or parents that they have a gene for a genetic disease? What if a family member does not want to know? How does genetic information affect planning for the future? Will the medical benefits be available to all or to only the wealthy or well insured? Will someone abuse this knowledge and create “super humans”? Should employers or insurance companies have access to genetic profiles? Could employers and insurance companies use genetic information to select their employees and insurers? Because the availability of treatment of genetic disorders remains many steps behind the diagnosis (Collins, 1999; Cunningham, 2000; Fink & Collins, 2000), why would one want a diagnosis or label for a condition for which no treatment exists? Even though many of these questions cannot be answered clearly at present, in their encounters with clients occupational therapists need to understand and be sensitive to the possible ethical dilemmas that their clients may be facing.

Nussbaum, McInnes, and Willard (2007) suggested that the principles of beneficence (doing good for the patient), nonmaleficence (do no harm), respect for individual autonomy (safeguarding a person’s right to control his or her medical care and medical information), and justice (ensuring that all individuals are treated equally and fairly) should serve to guide medical professionals when complex ethical issues arise. These principles, also found in the Occupational Therapy Code of Ethics (American Occupational Therapy Association [AOTA], 2005), should be familiar to all practicing therapists. As occupational therapists find themselves as part of medical teams, dealing with an increasing amount of genetically related ethical issues, they must be ready to work in collaboration with others to understand the client’s needs and requests, balance client choices with conflicting demands, and appraise each situation on the basis of established ethical principles.

Opportunities for Occupational Therapists

Although certain aspects of genetics research such as cloning and genetic engineering have drawn the most public attention, genetics will see its greatest impact through its effects on everyday life and in everyday health care. Therefore, occupational therapy professionals and other health care providers will play key roles in the use of genetics information in their practice (Kylar & Thomas, 2000; Lou, 2001; Schroeder-Smith, Tischenkel, De Lange, & Lou, 2001).

As clinicians, occupational therapists need to be prepared to understand genetic information as a client factor and to conceptualize genetic information as an overarching determinant of body structures and functions. Occupational therapists may be well placed to provide guidance for clients about the present and future impact of genetic disorders on participation in occupations. This would, however, require occupational therapy professionals to keep informed on fast-moving scientific developments. If occupational therapists develop and maintain a basic level of competence in genetics, they may be well suited to serve as part of genetic counseling teams, providing valuable insight with regard to function in daily living activities.

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In occupational therapy, understanding a client’s genetic makeup and predisposition to conditions can also prove valuable for treatment planning and developing intervention strategies (Kyler & Thomas, 2000; Lou, 2001). Occupational therapy professionals can tailor goals toward individual clients more effectively by recognizing a person’s predisposition for a particular condition and the potential impact on participation in future occupations (Schroeder-Smith et al., 2001). For example, when working with clients identified with susceptibility variants related to Type II diabetes, occupational therapists should be positioned to work with those clients in designing lifestyle plans for promoting healthy habits and routines (Jackson et al., 1998). For clients identified with susceptibility variants related to Alzheimer’s disease, occupational therapists should be working with them early on to use cognitive prostheses and to make environmental adaptations.

From a research perspective, occupational therapists are equipped to investigate one of the most important questions: Does knowledge of genetic predisposition result in an improved functional outcome for that person? From a psychosocial standpoint, occupational therapists may also ask how this genetic information affects motivation (e.g., to change behaviors), decision making (e.g., to pursue treatment or even access genetic information), and self-concept (i.e., how one views himself or herself and the future; Nussbaum, 2006). Occupational therapists could be valuable members of genetic research teams to help answer these questions, but we must seek these opportunities and argue that our professional knowledge would be an asset to these teams and valuable to the health care system as a whole.

Moving Forward, Taking Steps


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