Genetic Testing: Implications for Professional Nursing

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Scientific information emerging from human genome research has significant implications for the practice of professional nursing. Professional nurses assist individuals in making decisions about DNA studies, ensure that consent is informed before genetic testing, and help clients cope with genetic information after test results are known. Nurses with advanced education in genetics identify and counsel people who are candidates for DNA testing. Gene identification can have beneficial as well as harmful outcomes. Education is needed to prepare nurses for new roles regarding genetic testing. (Index words: Decision making; Genetic counseling; Genetic testing; Informed consent; Risk identification). J Prof Nurs 14: 184-188, 1998. Copyright © 1998 by W.B. Saunders Company

ONE OF THE GOALS of the Human Genome Project is to map and sequence the 50,000 to 100,000 genes in the human genome by the year 2005 (Andrews, Fullarton, Holtzman, & Motulsky, 1994). When a gene has been identified, it is possible to detect that gene in any individual before recognition of clinical symptoms of a disease regardless of the individual's age. Furthermore, once a gene has been mapped and sequenced, its relationships to physiological processes and interactions with environmental influences can be studied. Although all diseases result from an interaction between an individual's unique genetic makeup and the environment, in certain diseases the genetic component is so overwhelming that it expresses itself in a predictable manner (Goldstein & Brown, 1994). Although identification of the genes in the human genome is ahead of schedule, development of new treatments for genetic diseases and understanding of potential adverse personal and social consequences of gene detection are proceeding at a much slower pace.

Nursing care for people with genetic conditions traditionally has been associated with expectant families and children with birth defects. However, the identification of genes associated with adult-onset diseases, such as some forms of Alzheimer disease or familial breast cancer, makes it clear that knowledge of issues in gene identification is an essential part of professional nursing practice regardless of client age. Professional nurses may use several interventions with clients considering genetic testing.

Decision-Making Support

Decision-making support is the provision of information and support for a patient who is making a decision regarding health care (McCloskey & Bulchek, 1996). Professional nurses may provide decision-making support to individuals regarding genetic testing for the purpose of detecting a disease gene before onset of symptoms or to predict if the person is at risk to pass a disease gene on to his or her offspring.

Some insights into the decision-making process come from studies of people making decisions regarding having a presymptomatic genetic test for Huntington disease (HD). Expectations of people requesting presymptomatic testing for HD include wanting the information for family planning and a desire to reduce the uncertainty of not knowing whether one will develop HD. The most common reason cited in a study of 70 people was a desire to make plans for one's future (Tibben et al., 1993a). However, a longitudinal study of people undergoing HD testing showed that predictive testing may result in negative as well as positive consequences regardless of test outcome (Codori & Brandt, 1994). In this study of 68 people, the majority reported feeling relief from worry and uncertainty after learning their test results. Those who did not have the gene were consoled by the knowledge that their children were spared. Many of those with the gene experienced greater family closeness and were able to make plans for their future care. A negative consequence in those who did not have the gene was disappointment that this knowledge did not alleviate problems experienced throughout their lives. Psychological burden was the major negative outcome for those with the gene, and some persons in both groups reported a negative impact of gene detection in their

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marriages. Investigators note that presymptomatic testing for HD carries with it a significant risk of serious psychological distress and maladjustment, especially in those found to have the gene (Tibben et al., 1993b).

Some similar experiences were found in people requesting carrier testing for serious autosomal recessive or X-linked recessive conditions. Benefits included the ability to proceed with family planning decisions and a relief from fear that they could have been carriers. Individuals who learned that they were not carriers were relieved that their children were not at risk to be carriers (Williams & Schutte, 1997). However, for those who learned that they were carriers, burdens of this knowledge included feelings of sadness and loss of hope for healthy children. Difficulties in communicating results to some family members were experienced by both carriers and noncarriers.

In addition to the personal and family meanings of gene identification, results also may have social consequences, such as discrimination. Case studies of individuals having gene testing document discrimination in obtaining insurance, finding or retaining employment, and interacting with adoption agencies (Billings et al., 1992). The potential for discrimination is one consequence to be considered by people regarding gene identification. Research is needed to clarify factors influencing decision making regarding genetic testing and to guide nursing interventions for these individuals and their families.

Safeguarding Informed Consent

Genetic information may carry more risks and burdens than many other kinds of health information (Scanlon & Fibison, 1995). Thus, the informed consent process for genetic testing includes a discussion of benefits and risks of the testing; issues of disclosure of test results to others, such as family members or third parties; and subsequent decisions, such as reproductive decisions, that may be likely after results are known (Table 1). Professional nurses participate in safeguarding the informed consent process when genetic testing is offered for clinical as well as for research purposes. If the testing is offered on a research basis, additional information is needed, such as how the confidentiality of the specimen will be maintained, who has ownership of the specimen, and what is the person's future access to information that may emerge from ongoing studies (Weir & Horton, 1995). These issues are new and are not well understood by the public. Strategies for educating the public as well as persons faced with decisions regarding genetic testing need to be developed. Studies that clarify the impact of specific educational strategies on the understanding of genetic testing information are needed.

Coping With Genetic Information

Once gene identification is completed, some clients will need assistance in coping with the implications of their test results. A stress and coping model suggests that long-term distress from genetic testing results may be more likely to occur when the test suggests a high risk of illness, when a person's uncertainty regarding future health is not reduced, or when results of testing are in conflict with previously held beliefs. The lack of strong social support or useful coping skills are also believed to contribute to poor adjustment (Baum, Friedman, & Zakowski, 1997). Understanding a client's appraisal of the extent to which genetic testing may reveal harmful information as well as the client's appraisal of available resources are components of the professional nurse's assessment of a client's coping abilities. Knowledge of coping theory is an important component of professional nurse preparation for providing coping support to individuals undergoing genetic testing.

Gender is one factor that may influence the coping process. Follow-up studies of people having carrier testing for cystic fibrosis (CF) note that women are more likely to feel relieved about test results than men, suggesting that there may be gender differences in coping with the threat of being a carrier of a serious genetic disease (Marteau, Dundas, & Axworthy, 1997). Vernon et al. (1997) also noted a relationship between gender and psychological distress among people hav-
ing genetic testing for hereditary colon cancer. In their study, female gender, as well as less formal education and fewer and less satisfactory social contacts, were associated with increased distress. Coping is a complex process. These studies suggest that specific personal factors may be related to a person's abilities to cope with the results of genetic testing. Additional research may clarify the relationships between individual traits, social factors, and coping abilities.

**Maintaining Genetic Privacy**

Genetic information is uniquely personal and private. It can predict a person's likely medical future, it has a history of being used to stigmatize individuals, and it divulges information about one's parents, siblings, and children (Annas, 1995). When genetic testing occurs, the results may reveal information about one's family that the person does not want to know. One type of family information is misidentified paternity. When genetic data on several family members is used in test result interpretation, it may become apparent that the person identified as the father in a family is not the biological parent. In some testing centers, people are asked before beginning the genetic testing process if they want to be informed of this finding should it be present.

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Because genetic testing of one person can uncover information about other family members, maintaining privacy of genetic test results requires careful vigilance. Relatives may want to know the results of a person's test because it may help clarify their own gene status. However, the obligation to keep information in confidence is a duty that is basic to the health professions (Scanlon & Fibison, 1995). When results of genetic testing have implications for other family members, the nurse should ascertain what the client's wishes are regarding disclosure of test results to others.

**Risk Assessment and Genetic Testing**

Risk assessment for inherited conditions and obtaining genetic tests may be provided by advanced practice nurses who have additional education on genetics topics. Gene identification can be done to identify carrier status for some genetic disorders, to make a presymptomatic diagnosis of specific conditions, and to determine predisposition or susceptibility to developing certain disorders. Gene tests also are used to confirm a clinical diagnosis.

Carrier, or heterozygote, testing identifies whether a person has a mutation in one autosomal recessive gene or, in females, one gene on an X chromosome. When a person has only one copy of these genes, that person is a carrier. Carriers generally do not have clinical symptoms, but they could have a child with the disease if their child inherits the autosomal recessive gene from both them and their partner. CF is an example of an autosomal recessive condition in which genetic testing can identify whether or not a person is a carrier of a mutation in the CF gene. People with the highest risk to be carriers are those who have a family member with CF or who are white. The carrier frequency for whites of Northern European ancestry is 1 in 25, which is higher than in other ethnic groups. However, mutations in the CF gene have been identified in ethnic groups other than whites (Consensus Development Conference Statement, 1997).

Ten years before the Scanlon and Fibison (1995) report on nursing practice and genetics, the CF gene was identified (Tsui et al., 1985). Since that time, more than 600 mutations have been identified on this gene. Most people request carrier testing because they want to know their chances of having a child with CF or the chance that their children could be carriers of the gene. In one study of adults requesting carrier testing, the majority of participants learned of the test from a genetic clinic (Williams & Schutte, 1997).

Gene identification also can be used for presymptomatic diagnosis. In this instance, the test can document the presence of a gene for a clinical disorder before the onset of clinical symptoms of the disease. One example is the autosomal dominant disorder, HD. A person who has an ancestor with this condition is at risk to have inherited the gene. In this condition, the gene is virtually 100 per cent penetrant, meaning that a person who has the gene will develop the disease. However, no conclusive information can be derived from gene identification to predict age of onset or presenting symptoms, their severity, or their rate of progression. At the present time, predictive testing for HD only indicates whether or not someone has inherited the gene (Committee, 1994). In one study, people requesting presymptomatic testing for HD reported learning of the test from a genetic clinic,
from a relative, or from the media. None learned of it from their primary care provider (Holloway et al., 1994). Counseling and education in a specialized center are recommended whenever a person considers having a genetic test for conditions before the presence of clinical signs of the condition (Andrews, Fullarton, Holtzman, & Motulsky, 1994).

Predisposition or susceptibility testing is the third type of screening for gene identification. In this testing, genes are sought that convey an increased likelihood that a person will develop the disease. However, the likelihood is not 100 per cent. For example, the gene for the apolipoprotein (APOE) e-4 has been found to be associated with an increased likelihood that the person will develop Alzheimer disease (Corder et al., 1993). However, the presence of the gene does not mean that the person will definitely develop this disease, nor does the absence of the APOE e-4 gene mean that the person is free from the risk of developing Alzheimer disease. At this time, susceptibility testing for Alzheimer disease based on the APOE e-4 gene is not recommended.

Susceptibility testing is possible for some people who have a family history of hereditary breast cancer. If a woman is found to have one of the genes for breast cancer, the BRCA1 gene, the probability that she will develop breast cancer by age 70 is 87 per cent, and her risk to develop ovarian cancer is 44 per cent (Ford et al., 1994). However, test results cannot be used to predict when malignancy may occur. Furthermore, treatment such as prophylactic mastectomy may not completely eliminate the occurrence of cancer (National Cancer Institute, 1995).

Advanced education that includes basic genetic principles and the implications of genetic tests will be essential for nurses who will identify people at risk to have a gene for an inherited disorder. Most individuals receive genetic testing through specialty services, but some may undergo testing provided through their primary care provider.

Awareness of laboratory issues also is important when nurses participate in obtaining genetic tests. Genetic tests are provided by laboratories that have capabilities to provide tests for only one or several specific genes. Some laboratories provide testing as a part of research, whereas others offer testing on a clinical basis. Because some are commercial laboratories while others are associated with academic institutions, services such as cost, time required for test results, manner in which results are reported, and availability of professionals who can provide interpretation of test results varies considerably. In addition to laboratory personnel, other health professionals with expertise in genetics may be helpful regarding genetic testing issues (Table 2).

### Professional Nursing Preparation and Genetics

Genetic testing is a complex process, and nurses at all levels of practice will benefit from education regarding genetic testing. Formal educational opportunities in genetics are available in master's-level nursing programs (Table 3). Graduates of these programs function as advanced practice nurses with specific expertise in genetics. Many other nurses, such as advanced practice nurses in oncology, use knowledge of genetics in their own specialty area of practice. Implementation of a core curriculum in basic genetics for the oncology nurses is one strategy to address this education need (Jenkins, 1997). Integration of genetics into baccalaureate education for professional nurses can provide the foundation on which these nurses can incorporate genetic information as it becomes part of professional nursing practice. Introduction of genetic content for baccalaureate nurses is needed both in basic science and in clinical portions of nursing courses.

### Table 2. Genetic Resources for Professional Nursing Practice

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<tr>
<th>Resource</th>
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<tr>
<td>International Society of Nurses in Genetics, 3020 Javier Rd,</td>
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<tr>
<td>Fairfax VA, 22031; (703) 688-7355</td>
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<tr>
<td>National Society of Genetic Counselors, 233 Canterbury Dr, Wallingford,</td>
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<td>PA, 19086-6617; (610) 872-7608</td>
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### Table 3. Graduate Genetic Nurse Specialist Programs

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<tr>
<td>University of Cincinnati, College of Nursing and Health, 3110 Vine St,</td>
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<tr>
<td>Cincinnati, OH 45221; (513) 558-5380</td>
<td></td>
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<tr>
<td>Rush University, College of Nursing, 600 South Paulina St, Armour</td>
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<tr>
<td>Academic Center, Room 1022, Chicago, IL 60612; (312) 942-7100 (Admissions)</td>
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<tr>
<td>The University of Iowa, College of Nursing, 356 Nursing Bldg, Iowa</td>
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<td>City, IA 52242; (319) 335-7079</td>
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Summary

Professional nurses support decision making, safeguard informed consent, maintain privacy of genetic information, and support coping in individuals having genetic testing. With advanced education, nurses assess risk for genetic disease and participate in counseling regarding genetic testing decisions and results. Research is needed to identify factors associated with the education of clients about genetic testing and the impact of genetic testing on coping and overall health. Research also is needed to identify the effectiveness of nursing interventions to address problems encountered by people at risk for genetic conditions. Education of nurses on genetics at all education levels is essential for professional nurses to meet these goals.

References


