

Why Should Oncology Nurses Be Interested in Genetics?

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The Human Genome Project began in 1990 as an international effort to characterize human genetic instructions (the human genome) by creating a genetic map that reflects the position of genes on chromosomes. Scientists have completed a draft map of the human genome (International Human Genome Sequencing Consortium, 2001; Venter et al., 2001). Over the next decade, work will continue with computer technology to further identify genes associated with disease and the potential for interventions in risk reduction or targeted therapeutics.

In response to these advances, the National Cancer Institute (1998) established the Genome Anatomy Project to identify all the genes responsible for cancer development and malignant transformation. In the field of oncology, this project signals a dramatic shift in the way that patients will be screened, diagnosed, and treated. Oncology nurses will be required to know about genetics to understand the basic etiology of cancer.

The most simplistic definition of cancer is uncontrolled cell growth. People familiar with the discoveries of the Human Genome Project now understand that cancer is clearly a genetic disease. Genes are units of deoxyribonucleic acid (DNA). The genes code for normal proteins, which regulate cell growth. Damage (mutation) frequently occurs during normal cell division or as a result of environmental influences. When genetic damage escapes the normal repair mechanisms of the body, the mutations accumulate, resulting in uncontrolled cell growth. Most cancers occur because of multiple mutations involving several genes at each step in the carcinogenic pathway. Oncology nurses know that the carcino-

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genic pathway involves five steps: initiation, promotion, progression, invasion, and metastasis. The identification of the genes related to each step in the pathway will have a dramatic impact on oncology interventions. Thus, the increasing understanding of the genetic basis of cancer and the effect that understanding will have on treatment modalities will sweep oncology nursing into a new healthcare paradigm (Engelking, 1997).

The Impact of Advancements in Genetics on the Oncology Nurse's Role

Advances in genetics will influence every aspect of the cancer continuum, from prevention and screening to treatments and palliation. Oncology nurses, because of their holistic approach to patient care, have the opportunity to incorporate these advances into their role at each point along the cancer-care continuum. They can integrate genetics concepts into counseling, education, preparation of clients for decision making, and direct caregiving. Figure 1 shows the carcinogenic pathway and corresponding genetic events and nursing actions. The figure suggests ways that oncology healthcare

professionals can incorporate genetics principles into all aspects of patient care.

The field of cancer prevention is already focusing on the interaction of genes and environmental factors. Genetic profiles (such as blood type and human leukocyte antigen, or HLA, type) help to identify the individuals most susceptible to car-

cinogens. Genetic profiles are not profiles of mutations but of simple variations in genetic makeup. These variations, known as polymorphisms, often influence specific enzyme activity. For example, carcinogens present in tobacco are modified or detoxified by enzymes in the cytochrome P450 (CYP) family. Some individuals, by virtue of their genetic makeup, have a variation in one of the CYP genes and are unable to produce an enzyme capable of carcinogen detoxification. These individuals, both smokers and nonsmokers, are at higher-than-average risk for lung cancer (Bennett et al., 1999).

Oncology nurses working in the areas of prevention and risk reduction will routinely make use of genetic information to

- Identify high-risk populations through genetic testing or genetic profiles.
- Educate a high-risk individual about the effects of exposures based on the individual's genetic makeup.
- Recommend risk-reduction strategies that include lifestyle and behavior changes.

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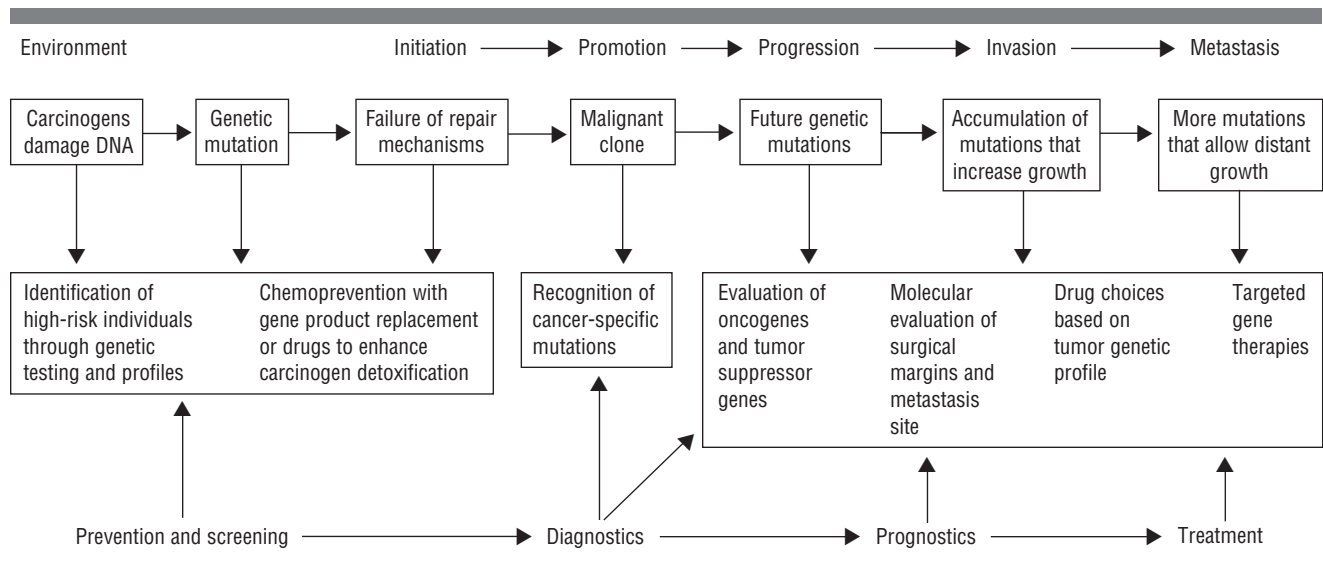


FIGURE 1. THE CARCINOGENIC PATHWAY, GENETIC EVENTS, AND CORRESPONDING NURSING ACTIONS

Note. Based on information from Peters, Dimond, & Jenkins, 1997.

- Counsel individuals about the benefits and limitations of genetic testing.
- Advise individuals about the psychosocial ramifications of genetic information. Some oncology nurses are specializing in cancer genetic-risk counseling. The technology to identify mutations in cancer-predisposition genes is clinically available for a variety of familial cancer syndromes, such as familial adenomatous polyposis (caused by an *APC* gene mutation), hereditary breast and ovarian cancer (*BRCA1* or *BRCA2*), hereditary nonpolyposis colon cancer syndrome (*MLH1*, *MSH2*), and multiple endocrine neoplasia 2a and 2b (*RET*). Oncology nurses working in cancer genetic-risk assessment and counseling
 - Educate individuals about the genes that predispose them to cancer.
 - Provide in-depth cancer-risk assessment by evaluating family histories and statistical risk estimates.
 - Guide individuals in decision making relative to genetic testing.
 - Prepare individuals and families for the potential impact of genetic information.
 - Address ethical concerns related to genetic information.
 - Advise individuals about the benefits and limitations of risk-reduction measures, such as chemoprevention, prophylactic surgery, and lifestyle changes.

Nurses involved in the clinical arena are already using the terminology of genetics when they explain the diagnostic and prognostic features of cancer cells. As genetics continues to advance, the accuracy of diagnosis and prediction of treatment response will increase. Nurses will need to understand and explain

- Which genetic features, such as the absence or presence of a gene product or genetic changes noted histologically (Quirke & Mapstone, 1999), help to identify early cancers
- Molecular staging, which predicts which patients are at high risk for disease spread (For example, molecular staging can help to identify genetic lesions in surgical margins—lesions that are not yet malignant but are indicative of residual disease [Brennan et al., 1995].)
- How genetic markers in peripheral blood or body fluids are predictive of disease spread or relapse (Kodera et al., 1998)
- How genetic characteristics of the tumor can predict the patient's response to chemotherapy.

Innovative therapies will use replacement genes or gene products to treat cancer. Tumor cells will be genetically modified to make them more susceptible to conventional therapies. Oncology nurses will have to understand the genetics behind these soon-to-be commonplace practices.

Public interest is high regarding the use of genetic information to predict cancer susceptibility (Lerman, Daly, Masny, & Balshem, 1994; Smith & Croyle, 1995). The promise of better cancer prevention and treatment has created consumer demand for genetic information and genetic services. As a result of this demand, oncology nurses have been involved in genetic predisposition testing—that is, identifying and counseling carriers of an inherited mutation (Calzone, 1997; Giarelli, 1997). Diagnostics have always preceded therapeutics in every major paradigm shift in healthcare practice (Col-

lins, 1997). For example, the identification of bacteria was a diagnostic discovery that was well in advance of antibiotic therapies. In the same manner, genetic testing is the diagnostic precedent to major changes in oncology practice. Figure 2 shows the steps from the identification of genes involved in disease to the development of therapies. Note the time gap between the detection of contributing genes and the emergence of new treatment modalities. In the field of cancer genetics, computer and DNA technology are expected to make this gap a short one. Therefore, the window of opportunity for oncology nurses to incorporate genetic information into their roles as educators, counselors, and primary caregivers is now.

Genetic Health Care and Oncology Nursing

Genetic health care is care for people whose health, wellness, or disease is caused or influenced by genes. Healthcare services must integrate genetic health care into the continuum of cancer care.

Discoveries about how genes direct the construction and operation of the human body are showing how genetic changes influence disease susceptibility and development. These discoveries offer opportunities for improvements in the care of individuals with cancer through the integration of genetic concepts into all healthcare visits.

Using genetics technology in the clinical setting creates challenges for both healthcare providers and consumers. The interest, the technology, and now the expanding

GENE-MAPPING TIMELINE

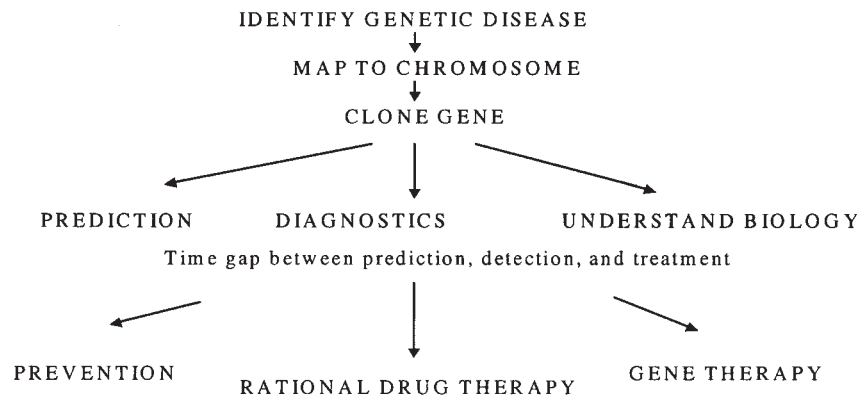


FIGURE 2. THE STEPS INVOLVED FROM THE IDENTIFICATION OF GENES INVOLVED IN DISEASE TO THE DEVELOPMENT OF THERAPIES

Note. From "Clinical Applications of Genetic Technologies to Cancer Care," by J. Peters, E. Dimond, and J. Jenkins, 1997, *Cancer Nursing*, 20, p. 365. Copyright 1997 by Lippincott Williams and Wilkins. Reprinted with permission.

applicability to all individuals with cancer provides opportunities for nurses to design new genetic health services that enhance outcomes for individuals and families. The opportunities also include different and difficult choices that accompany genetic information.

How Is Genetic Information Different?

Soon it will be possible to determine a person's individual genetic profile and his or her risk of specific disease. Information about a person's genetic makeup is very personal and reflects a permanent part of that individual. At the same time, every individual's genetic information has a familial component in the sense that genes convey traits, including cancer risk, from generation to generation. A patient's blood relative may carry the same genetic information the patient carries.

The fact that genetic information pertains to an individual and to his or her family raises issues regarding individual versus family rights to confidentiality and the right of each person to know versus the right not to know about genetic factors. Ethicists have debated whether individuals in a family have the right to be informed about genetic factors that could affect their well-being and choices (Parker, 1995; Van Leeuwen & Hertogh, 1992). For example, should healthcare providers share information about misinformed paternity? Do family members have the right not to know about cancer risk? Will

knowledge of predisposing genes lead to pressure from a healthcare provider or third-party payor to modify lifestyle and screening practices? Will knowledge of genetic markers that family members share disrupt or strengthen family relationships? A better understanding of how individual uniqueness affects familial factors would help to answer these questions.

Although a person's genetic makeup is permanent, whether his or her makeup will result in undesirable consequences is uncertain (Lerman, 1997). This uncertainty affects personal decision making, which also is influenced by prior perception of risk, values, attitudes, and cultural beliefs. Identifying people who have a predisposition to developing an illness may lead asymptomatic people to question their own concept of being healthy.

Genetic information often provokes emotional and behavioral responses. It could affect insurance coverage and employability and cause discrimination.

To summarize, genetic information is different from other health-related information because it could reflect family members' risks, it does not reflect certainty about whether risk will lead to disease, it provokes unusually emotional responses that affect behavior, and it has a very high potential to affect insurance coverage and employability and expose the client to discrimination. Because of these unique features, healthcare professionals must handle genetic information with greater consideration than is applied when handling other health-related data, such as blood counts. In addition, be-

cause of the unique nature of genetic information, healthcare professionals must ensure informed consent before incorporating genetic information into general healthcare and nursing practices.

How Will Genetic Information Be Incorporated Into General Health Care?

The discovery of the contributions genetics makes to health and illness will affect the care of all clients (both adults and children) at all clinical settings and across all practice specialties.

In terms of cancer care, nurses can present information about genetics to clients during the initial assessment; during discussions of early detection, cancer prevention, diagnostics, medical surveillance, and the causes of different types of cancer; and when planning risk reduction, predicting prognosis, designing treatment options, and monitoring disease response.

What Role Will Nurses Play in Meeting the Genetic Healthcare Needs of the Future?

With some augmentation of the education they already have, nurses will be able to provide the counseling and teaching that recent genetic discoveries necessitate. Nurses must plan proactively to build on that foundation by enabling personal skill development and ensuring that they continually update their knowledge of genetics by learning about ongoing scientific discoveries that affect client care. Translating science into practical terms has always been a nursing priority. In the fast-changing field of genetics, nurses will again find ways to apply exciting scientific discoveries to the care of individual clients. This text will provide examples of responsibilities to consider in the design of a personal plan to meet these challenges.

Nurses have an opportunity to provide leadership in the design of genetic healthcare services that offer safe and ethical applications of genetic technology. Nursing's role will be important in the assessment, planning, implementation, and evaluation of cancer-genetics health services.

For oncology nurses, the integration of cancer genetics into oncology care presents countless opportunities for personal and professional growth. With these opportunities

comes the obligation to ensure that genetic information and technology enhance cancer care. Another obligation is to consider the ethical, social, and legal implications of genetics knowledge along with the medical application of cancer genetics technology.

A brochure published by the American Nurses Association (ANA), *Managing Genetic Information: Implications for Nursing Practice* (Scanlon & Fibison, 1995) may help nurses to integrate ethical principles into nursing guidelines regarding genetic information.

Patient advocacy, nursing research, and legislative efforts offer additional opportunities to influence healthcare policy in regard to applying genetics to clinical care. Understanding the implications of genetic information for the individual, family, and society will enhance the ability of nurses to influence decisions that affect practice, education, and the quality of cancer care. The Oncology Nursing Society (ONS) has made the commitment to prepare its members to meet the challenges created by the Genetics Revolution (ONS, 1997). The publication of this text is one ONS-sponsored initiative designed to help to prepare nurses to incorporate genetics into practice. This text will define the education that nurses will need to accomplish this task. It will establish standards and guide practice in cancer genetics and will help to define the role of the nurse in genetic health care. This text will serve as a resource for nurses presented with genetics issues in their practice, and it will provide a model for nurses as they incorporate genetics into their practice.

Are You Using Genetic Information Yet?

You may be surprised at the extent to which you already incorporate information about genetics into your practice, even though you may have no formal training in genetics.

An ANA survey showed that only 9% of nurses in the study had genetics training. Most (68%) indicated that they were not at all or not too knowledgeable about genetics. However, most nurses reported performing some genetics-related activity at least occasionally, with a majority citing low levels of confidence in explaining genetic information (Scanlon & Fibison, 1995).

You may be performing genetics-related activities and using genetics information without recognizing it. To see how often you use or provide genetics information, take the quiz that follows. The quiz will help you to assess your knowledge of genetics. The material that follows the quiz will present the answers.

Quiz: How Frequently Do You Use or Provide Genetic Information?

Mark a check on the line that appears before “Yes,” “No,” or “Not sure” to answer the questions that follow.

- 1a. Have you ever told individuals to use sunscreen?
___ Yes ___ No
- b. Do you know the genetic basis of this recommendation?
___ Yes ___ No
- 2a. Has a patient ever told you about other family members with cancer?
___ Yes ___ No
- b. Do you understand the genetic basis of their concern?
___ Yes ___ Not sure
- 3a. Have you ever explained to a patient how tumor markers function in a workup or cancer follow-up?
___ Yes ___ No
- b. Is it true that some tumor markers are genes expressed by tumors?
___ Yes ___ Not sure
- 4a. Have you ever seen the term *aneuploidy* on a pathology report?
___ Yes ___ No
- b. Did you know that *aneuploidy* refers to DNA changes?
___ Yes ___ Not sure
- 5a. Have you heard of vaccines for melanoma?
___ Yes ___ No
- b. Did you know that treatment by means of a vaccine is a form of gene therapy?
___ Yes ___ Not sure
- 6a. Do you know why insurance companies ask about family history of heart disease or cancer?
___ Yes ___ Not sure
- b. Do you know the social implications of the answer to the preceding question?
___ Yes ___ Not sure

Scoring

To see how often you use genetics information, count the number of times you answered “yes” to questions labeled a.

- 0–1: You seldom use genetics information.
- 2–3: Like most nurses, you occasionally use genetics information.
- 4–6: You already use genetics information frequently in your nursing practice.

To see how much knowledge you have to explain genetics information, count the number of times you answered “yes” to questions labeled b.

0–3: Like most nurses, you have a basic understanding of genetics but are willing to incorporate new information into practice.

4–6: You already have a foundation of knowledge about genetics upon which you can build.

Quiz Answers

1. The recommendation for sunscreen has a basis in genetics. Researchers believe the pathophysiology of cancer is related to the interaction of genes and the environment. Multiple exposures to ultraviolet light cause genetic mutations that can lead to skin cancer.
2. When patients tell you about family members with cancer, they may be concerned about the impact of family history on the potential for disease in themselves or a family member. The media have focused attention on the potential hereditary aspect of some cancers. From 5%–10% of all cancers have a hereditary component.
3. Tumor markers are proteins, antigens, enzymes, or genes expressed by the tumor or produced by normal tissue in response to the tumor. For example, the *MYC* oncogene *ERBB2* (HER2-neu) is a genetic tumor marker used in the diagnosis and monitoring of cancer.
4. DNA analysis of solid tumors characterizes the DNA as normal (diploid) or abnormal (aneuploid). Aneuploidy is abnormal or disorganized DNA. DNA analysis helps to determine prognosis by assessing the proliferative potential of tumors.
5. Vaccines are a type of gene therapy, an approach to cancer treatment that falls into the class of immunotherapy. Vaccines stimulate the immune system’s ability to mount a response against a cancer. A cancer vaccine is designed to immunize patients against their own cancers by injecting them with their own tumor cells after the cells have been modified by certain genes. The goal is to make the cancer more sensitive to chemotherapeutic agents.
6. Family history currently is included as one of the classifications of genetic information. Potential discrimination for health or life insurance is an ongoing concern in cases in which the covered person has a family history of cancer.

The Kennedy-Kassebaum legislation, enacted July 1, 1997, provides some protection against health insurance discrimination for preexisting genetic conditions for people already insured by a group plan. Federal workers are covered by an executive order that states that federal employers may not use genetic information to deny employment, make job assignments, or guide promotion decisions (National Human Genome Research Institute, 2000). The order also limits the ability of federal employers to collect or disclose genetic information about an applicant or employee. No legislation protects individuals from genetics-based discrimination regarding life insurance.

Summary

Your answers to the quiz probably showed you that genetics is already part of your oncology nursing practice. Without realizing it, every nurse practicing in oncology is already using genetics information in the context of promoting healthful behaviors to prevent cancer; identifying people at risk for cancer; explaining features of cancer that affect prognosis and treatment; and addressing the legal, social, and ethical impacts of cancer.

The work of the Human Genome Project and the mapping of protein-coding genes are helping to unlock the molecular basis of the initiation, invasion, progression, and metastasis of cancer. These findings will continue to elucidate the understanding of every aspect of the cancer continuum and will initiate advances with the potential for novel interventions in risk reduction and targeted therapeutics.

This text will help oncology nurses to understand the genetic basis of cancer. Although this text focuses on assessment, it will help readers to explore how genetics will influence the entire cancer care continuum. The authors anticipate that our growing knowledge of cancer genetics will serve to further the mission of ONS: "to promote excellence in oncology nursing and quality cancer care."

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