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The Research Connection

The Psychosocial & Nursing Advisory Board to the New Jersey Commission on Cancer Research

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The Joint Psychosocial & Nursing Advisory Group to the NJCCR was appointed to advise the Commission of special research needs pertaining to nursing, psychology, sociology, and related disciplines for the purpose of addressing gaps in vital areas of cancer research and cancer care in New Jersey.

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Introduction

by
Kathleen Neville, Ph.D., RN
Professor, Department of Nursing
Kean University

For the past two years, the Journal of Nursing Scholarship, the official publication of the Sigma Theta Tau International Honor Society of Nursing has published numerous articles in a series focusing on Genomics for Health. This attention clearly demonstrates the current and future influence of genomics on health prevention, disease prevention, and diagnostic and treatment approaches (Prows, Glass, Nicol, Skirton, & Williams, 2005). In earlier years, health professionals might have viewed genetics as a discipline that had particular relevance to the pediatric population with less relevance to other age groups. However, genetic disorders can be manifested at any point in one's life and there are no age, social, economic, ethnic or religious barriers (Lashley, 2005, p. 3).

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Scientific progress in genetics and genomics has transformed traditional health care, not only in cancer, but in all health conditions. While all cancers are believed to have an underlying genetic basis, the effect of environment and lifestyle are strong influencing factors. Approximately 5-10% of malignancies are believed to be part of a hereditary syndrome, whereby usually in these families, more than one family member develops cancer (Calzone, Lea & Masny, 2006). Greater understanding of these critical factors and their interrelationships has and will continue to significantly influence the delivery of care and ultimately health outcomes.

In recognition of the need for the U.S. nursing workforce to become proficient in genetics and genomics, in 2004, the National Human Genome Research Institute and the National Cancer Institute began a broad training initiative (Jenkins & Calzone, 2007). As an outcome of this initiative, specific competencies have been developed and have been endorsed by leading nursing organizations nationwide. Additional information about this initiative can be obtained at http://www.nursingworld.org/ethics/genetics.

Using a framework of genetics in the cancer care continuum, Lea, Calzone, Masny & Bush (2002), address the genetic influence of nursing interventions along the cancer care continuum. In this framework, the influence of genetics (the study of single gene disorders) and genomics (the complex interface of the entire genome with environmental factors) on professional practice across the cancer care continuum is addressed. Ranging from the beginning phase of conception, whereby health professionals focus on physical, family and lifestyle assessment, additional phases of the continuum are initiation to diagnosis, treatment/prognosis, disease progression and management of end of life. All of these phases require specific biological knowledge to guide interventions and practice.

Emerging genomic discoveries will continue to shape health care and impact on human health. Similarly, professional responsibilities and practice issues continue to evolve as the unprecedented progress in scientific and clinical research advances.

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ANNOUNCEMENT

A Resource Book for Cancer Patients in New Jersey
has been revised
by the Nursing & Psychosocial Advisory Group to the NJCCR.

Copies are now available, free of charge, by calling 609-631-4747.

The Use of Genetic Technology in Pediatric Oncology

by
Larissa Labay, Psy.D. and David Gordon, M.S.
Tomorrows Children's Institute
Hackensack University Medical Center

The pediatric oncology medical community has provided consistent, standardized medical care for virtually all children and adolescents with cancer. Almost uniformly fatal 40 years ago, pediatric cancer is now a frequently curable illness. In the US, nearly 13,000 new cases of cancer are diagnosed each year in children and adolescents less than 20 years old. The overall 5-year survival estimate of these patients is approaching and soon to exceed 80% (Reaman, 2004). One important reason for this progress is that approximately 75% of pediatric cancer patients are treated on a nationally standardized protocol. In practice, this means that children at different treating institutions regardless of geography, socioeconomic status, race, and other variables receive essentially the same medical treatment based on their specific childhood cancer diagnosis. This high level of medical intervention and support is one of the success stories in modern medicine.

Yet despite this high level of standardization, it is a fact that individual children treated on identical treatment protocols can have remarkably different treatment results and vastly different long term effects from their therapy. Current research in genetics is working toward better understanding this phenomenon and will lead toward more tailored treatments with less severe long term effects. It is known that 10 to 15% of all childhood cancers are hereditary (Quesnel & Malkin, 1997). This is significant as information from genetics can provide knowledge not just for the patient who has been diagnosed, but for other relatives who may be at risk of developing these same cancers.

Advances in genetic technologies make it possible for providers to tailor treatments to individuals. Large scale studies are showing that survivors of childhood cancers are at an increased risk of developing a secondary malignancy. Currently, genetics research is focusing on discerning who is at greatest risk of developing secondary malignancies as well as creating treatments of the future which will decrease or eliminate the risk of secondary cancers. Much hope lies with pharmacogenomics, where pharmacologic agents are matched with patients based on genetic analysis to improve treatment outcomes as well as decrease susceptibility to adverse short and long term effects (Ansari & Krajinovic, 2007).

While advances in the field of cancer genetics may have a positive influence on treatment regimens, it is important to consider the ethical and psychosocial ramifications of this technology. The American Society of Human Genetics and The American College of Medical Genetics (1995) issued a joint report regarding the ethical, legal, and psychosocial implications of genetic testing in children and adolescents. Timely medical benefits and substantial psychosocial benefits were identified as the primary justifications for genetic testing in the pediatric population. This report acknowledged that although it is presumed that parents have the best interest of a child in mind when making decisions regarding genetic testing, it is important for the practitioner to have a thorough understanding of the implications of such testing and serve as an advocate for children during the process. A major concern is that because the decision to have a child genetically tested is made by parents, it precludes that individual from making the autonomous decision whether or not to be tested as an adult. Therefore, practitioners need to consider the developmental stage of the child and determine the extent to which they should be involved in the decision making process, and attempts should be made to obtain assent when appropriate. Finally, it is important to determine whether there are disparities in access to genetic technologies that may improve outcomes for the pediatric oncology population.

Psychosocial implications associated with genetic testing include anxiety, fear about the future, guilt, and difficulties with self-esteem (AAP, 2001). Hypervigilance about health issues is common for pediatric oncology patients, survivors, and family members, and may be heightened in cases where hereditary cancers are identified (Patenaude & Schneider, 2006). This is particularly relevant because the familial nature of genetic

information can have medical and psychosocial implications for multiple family members. Such testing can also create tension between parents who struggle with feelings of guilt and blame after receiving information regarding the hereditary nature of cancer in the family (Patenaude & Schneider, 2006). Psychosocial challenges are also faced by survivors of hereditary cancers as they reach young adulthood and face decisions regarding reproduction and the possibility of risk to their children. Finally, because individuals with hereditary cancers are at an increased risk for second malignant neoplasms, many families worry about possible employment and insurance discrimination. Fortunately, the majority of states have legislation in place that protects the privacy of genetic information.

The complex ethical and psychosocial implications of genetic testing underscore the need for families to engage in a well informed decision making process. Ideally, decisions should be made in collaboration with knowledgeable genetic counselors who understand the implications of genetic technologies on both medical and psychosocial well-being. In response to these issues, it is standard practice for specialized programs in pediatric oncology and genetics to incorporate psychosocial support services to help families throughout the process of genetic testing.

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Legislative Update – Genetic Privacy

by
Ann Marie Hill
Executive Director
NJ Commission on Cancer Research

The discovery of BRCA I and then BRCAII in the early 1990s triggered considerable alarm by breast cancer survivors and advocates around a number of privacy and discrimination issues surrounding genetic testing. Fear that insurers and employers would obtain genetic test results and then try to restrict coverage for individuals with positive tests became a rallying cry for action. Concerns about employment discrimination and social stigmatism were also raised. As a result, a number of states, including New Jersey, began to pass laws to protect privacy and prevent discrimination for those interested in undergoing genetic testing.

The New Jersey Genetic Privacy Act (PL 96, C. 126) was signed into law on November 19th 1996 by Governor Christine Todd Whitman. At the time, it was heralded as one of the strongest laws regarding genetic privacy in the country. It regulates genetic testing, the retention and disclosure of information obtained from genetic tests, protection against the unauthorized use of genetic information and health insurance or employment discrimination on the basis of genetic information. The law also precludes unfair discrimination by the life insurance

industry based on genetic risk, a first at the time. It was also one of a handful of laws at the time that actually had penalties including potential prison terms for violations.

Most states now have some type of genetic privacy or discrimination law in place. According to the National Association of State Legislatures, forty nine offer some type of protection against discrimination for health insurance (Johnson, 2007). Thirty-three include some type of protection for life and disability insurance and thirty weigh in on different privacy issues. Different definitions, regulations and penalties are used by the individual states resulting in an inconsistent network of loose protections for individuals. Obviously, state laws offer protection only within the state's boundaries. In addition, states can only regulate certain types of insurance contracts and large self insured company (ERISA) plans are usually exempt from state oversight.

In the information age, a haphazard network of state laws does not offer protection to individuals or their family members. With the enactment of the Health Insurance Portability and Accountability Act of 1996, federal law finally began to address the privacy and discrimination issues. Health insurers cannot use "genetic information" in "granting, denying, canceling or otherwise evaluating an individual's eligibility for coverage" (OCR, 2003). The law specifically says that insurers cannot apply "pre-existing" condition rules to genetic information unless the person has been diagnosed with the disease already (Shay, 1997). While HIPPA itself does not address specific protections against discrimination based on genetic information, it does empower Health and Human Services (HHS) to do so. The so-called, "Privacy Rule" published by HHS in response to HIPPA provides a foundation of federal protections for individual health information including genetic information (Shay, 1997).

Still, federal legislation specifically focused on genetic discrimination and privacy issues has faced a stormy road over the years. Numerous bills have been introduced only to be passed by one house but not the other. However, the Genetic Information Non-discrimination Act of 2007 (GINA, H.R. 493) may well be law by the time this newsletter is printed.

Under GINA, group and individual health plans, and issuers of Medicare supplemental policies are prohibited from requesting or requiring an individual to take a genetic test. H.R. 493 extends medical privacy and confidentiality rules to the disclosure of genetic information in underwriting (presently permitted under HIPAA) and makes it unlawful for employers to discriminate against individuals in hiring, firing, job assignments and promotions based on genetic information (Slaughter, 2007). This prohibition is extended to unions, employment agencies and labor-management training programs.

On April 25, the House passed GINA by a margin of 420-3 and the act was moved to the Senate which has a similar bill (S398) that has already been placed on the Senate Legislative Calendar for a vote. The President has said he would sign this legislation if passed. H. R. 493/S. 358 amends the Employee Retirement Income Security Act of 1974 (ERISA), Internal Revenue Code (IRC C. 100) and the Public Health Service Act to expand the prohibition against discrimination by group health plans or insurers on the basis of genetic information. H.R. 493, but not S. 358, would amend HIPAA by restricting use and disclosure of genetic information (Slaughter, 2007).

With major advances in genomics, proteomics and bioinformatics, new technologies including the prediction and modification of cancer risk, molecular profiling for diagnostic and prognostic purposes and targeted therapies based upon pharmacogenomics, are rapidly moving forward. A willingness by individuals to participate in clinical trials will be essential if such advances are to reach the clinic. Finally, those individuals at risk of diseases such as cancer must feel free to take advantage of the emerging technologies without fear or harm. If GINA passes as expected, individuals should be more encouraged to take advantage of genetic testing and be more willing to participate in genetic research.

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Not just a blood test…

by
Kimberly Ranieri DeLeonardis, MS
Certified Genetic Counselor
The Cancer Institute of New Jersey

Genetic testing typically refers to the identification of changes in genes that are associated with an inherited predisposition to a condition or trait. Genes are present in most cells of the body; so genetic testing can be performed on various types of tissue, most commonly on a small blood sample. Genetic testing for hundreds of inherited conditions is commercially available with some labs even marketing directly to consumers. With the widespread availability of genetic testing, many of our patients are asking, "Why is genetic testing such a big deal? Isn't it just a blood test?"

As you are likely aware, genetic test results can have implications for an individual's medical management. Individuals found to carry a genetic predisposition to develop certain types of cancer may elect to alter their medical management strategies. Options available to them include increased surveillance, chemoprevention and/or prophylactic surgery. However, in most discussions regarding the impact of genetic testing, the psychosocial implications of genetic testing are often overlooked.

As healthcare professionals, every patient should be provided the opportunity to consider the psychosocial sequelae of genetic testing as well as the medical implications. Psychological topics that may be discussed with patients prior to genetic testing include, the patient's anticipated reaction to a positive or negative test result, the potential effect of test results on personal and family relationships, and the level of interest other family members have in genetic information.

The psychological impact of genetic test results will obviously vary from patient to patient, depending on their personal or family history of cancer, and their psychological state before genetic testing. Unaffected individuals with a positive genetic test may experience a psychological response similar to a cancer diagnosis itself. Initial feelings of anxiety, fear and apprehension may be accentuated by the uncertain risks associated with having a genetic predisposition, and the length of time between genetic testing and potential diagnosis (Pasacreta, 2003). An individual with a cancer diagnosis who receives a positive genetic test result may feel more in control, and less uncertainty regarding their future cancer risks (Claes, 2004). Once a mutation has been identified in a family, many patients voice concern about the cancer risks for their children in the future, whether the child is 2 years old or 42 years old. A patient's response to a negative genetic test result may vary between relief or feeling less in control of future cancer risk due to residual uncertainty of cancer risks. Any individual receiving genetic test results should be provided with a supportive environment to help them adjust to their test results, regardless of the outcome.

Any individual pursuing genetic testing faces the challenge of balancing the medical and psychosocial implications of genetic test results, not only for themselves, but also for their family members. Genetic testing is unique in that an individual's genetic test results may have implications for the entire family. Genetic test results can also alter family dynamics, as not all

individuals in the family are likely to perceive genetic information in the same way. Some family members may want genetic information to help manage their own medical care or lifestyle choices. Other individuals in the family may not choose to learn this information if they do not feel it will be beneficial to their medical care, or if they are concerned about other issues, such as insurance discrimination or privacy concerns. In families, some individuals will inherit a familial predisposition to develop cancer, while others may not. A family member with a familial cancer predisposition may feel jealous of unaffected family members or angry because they inherited the familial predisposition while others did not. A family member who does not inherit a familial cancer predisposition may feel guilty for feeling relieved about their own unaffected status, and experience a sense of survivor guilt. It is useful for patients to recognize potential issues that may arise when genetic test results are disclosed to family members.

Patients who are identified with an inherited predisposition should be encouraged to share this information with family members, who may also be at increased risk for certain cancers. However many patients may experience trepidation at the prospect of sharing this type of information with their relatives, for fear of being the bearer of bad news to their family members (Bonadona et al, 2002). Sharing personal genetic test results may jeopardize an individual's privacy, or introduce conflicting emotional responses into the family. On the other hand, sharing genetic test results may elicit emotional support, sharing of resources, and critical assistance in obtaining information and medical care. Patients should consider how they want to share this type of information with their family during the genetic testing process.

Genetic counseling is a process that incorporates genetic education, medical management and risk assessment with a discussion of the psychological issues related to genetic testing. Genetic counselors are health care professionals uniquely trained to provide this type of psychosocial perspective and support during the genetic testing process. Typically genetic counselors can spend 1-2 hours at a pre-test counseling session to make sure all of the potential implications of genetic test results are explored with a patient prior to pursuing genetic testing. Disclosures of all genetic test results in person provide the opportunity to assess the patient's psychological state, as well as their comprehension of technical medical information. Referring individuals interested in genetic testing to a genetic counselor gives patients the chance to discuss these issues prior to pursuing genetic testing.

Although one can never anticipate every situation that patients will encounter, the role of health care professionals is to facilitate the process of informed consent for each patient. Genetic testing carries unique stressors for patients, as well as for their families. It is important that patients are given the opportunity to carefully consider the implications of a genetic test on both their medical care and their psychological well-being, and to understand why genetic testing is "not just a blood test".

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ATTENTION

The NJCCR is planning to publish this newsletter via email only by the end of 2007.

Also, in order to offer our readers more, we plan to offer nursing, social work, and other professional CEU credits.

Therefore, please provide your name and contact information, most importantly your email, to Michael Toleno at (njccr@doh.state.nj.us).

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