
Incorporating Genetic Counseling Into Primary Care Practice

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The era of genomic medicine is upon us. The completion of the Human Genome Project has allowed researchers to identify new genes responsible for common diseases, such as cancer and diabetes. As a result of these new discoveries, primary care physicians will increasingly be called upon to arrange for genetic services for their patients. Many physicians are too busy or have too little training and/or interest in genetics. Genetic counselors have specialized training in medical genetics and counseling and can assist primary care physicians in meeting the growing demand for genetic services.

A new era of genomics is changing the practice of medicine. Genomics, a term coined more than 18 years ago,(1) describes the shift in focus from the study of a specific gene and its function (genetics) to the study of the function and interaction of all of the genes in the human genome.(2) Benefits to patients in pregenomic medicine were limited to ordering genetic tests to diagnose rare hereditary conditions, confirm a clinical diagnosis, or learn about the risks for occurrence or recurrence of the disease. The analysis and interpretation of genes and gene expression made possible as a result of sequencing the DNA of the entire human genome is revolutionizing the way in which we diagnose, treat, and, in some cases, prevent diseases. It is through disease prevention that the advances made by the Human Genome Project will have the greatest impact on human health and patient care.

Predisposition Genetic Testing

Predisposition genetic testing for hereditary cancer syndromes is a current area of focus in cancer prevention. The majority (49%-90%) of hereditary breast and ovarian cancers are caused by a mutation in 1 of 2 genes, BRCA1 or BRCA2.³ Women who carry a mutation on either gene face lifetime risks as high as 85% for breast cancer and 40% for ovarian cancer.⁽⁴⁾ Identifying women at increased risk for cancer before the disease occurs gives the primary care physician crucial information for developing cancer prevention strategies tailor-made to the individual patient.⁽⁵⁾

Knowledge of whether a woman who is affected with breast cancer also carries a hereditary BRCA1 or BRCA2 mutation is proving to be important in determining treatment protocols as well. A recently published study reported that women with breast cancer who carry a BRCA1 or BRCA2 mutation respond better to certain chemotherapeutic drugs than others.⁽⁶⁾

Predisposition testing for more common diseases, such as diabetes, atherosclerosis, or dementia, will soon be possible. These common conditions are the result of a combination of genetic and environmental factors and are inherited in a multifactorial manner. Further analysis and understanding of gene-gene interactions and the environmental influences on our genetic code will help create more accurate genetic tests, which, in turn, will likely increase the number of requests for predisposition testing. With approximately 60% of the population affected by diseases that are inherited in a multi-factorial fashion,⁽⁷⁾ there is little doubt that genetic services will become a routine component of most patients' medical care, and primary care physicians will frequently be asked to assume the responsibility for coordinating such services, if not actually providing them. A complete discussion of which tests are available and who to test is very complex and goes beyond the scope of this article.

Benefits of Genetic Counseling

Recent research has demonstrated the benefits of genetic counseling. A study of 37 adults with at least 1 first- or second-degree relative with colorectal cancer who underwent genetic counseling and DNA testing found that all but 1 who had a susceptible gene identified on genetic testing told their relatives about the testing and that relatives of 3 of the patients with a mutation requested testing for themselves.⁽⁸⁾ The participants who underwent genetic testing and counseling were also more likely to undergo recommended surveillance testing for colorectal cancer. A separate meta-analysis of 25 different trials found that genetic counseling for familial cancer appeared to have no adverse effects on general anxiety or cancer-specific worry.⁽⁹⁾

Cancer testing counseling

The American Society of Clinical Oncology recommends that all individuals undergoing genetic testing receive extensive pre- and posttest genetic counseling.(10) Our current understanding of the molecular basis of cancer makes it possible to identify hereditary cancer susceptibility genes in individuals with a strong family history of cancer. Patients who are screened by the primary care physician and identified as belonging to a family with a potential hereditary cancer syndrome (Table 1) should then be referred to a genetic counselor for a comprehensive cancer risk assessment.

Illustrative case

The following case exemplifies the genetic counseling process for a hereditary cancer predisposition syndrome.

A 29-year-old man tells his internist that he is concerned about his family history of gastric cancer. He reports that his sister was diagnosed with and died of gastric cancer at age 29, his father was diagnosed with the disease at age 37 and died at age 40, and his father's sister was diagnosed with and died of gastric cancer at age 40. In addition, his paternal grandmother, who is now 84, was diagnosed with breast cancer at age 62. The man has 2 daughters, aged 4 and 2.5 years, and he and his wife are expecting their third child in several months. He asks about his risk for gastric cancer and what prevention strategies he and his family can pursue. He also wants to know about the risk his children may have for this deadly cancer.

For this patient, a comprehensive risk assessment session with a genetic counselor begins by elucidating a thorough family history, which is constructed into a detailed pedigree. Using the family history, environmental exposure history, and medical history, a risk assessment for developing a specific cancer or for carrying a hereditary cancer gene (based on the patient's concerns and background) is provided. Depending on the types of cancer in the family, the risk assessment can also include a numerical estimate of risk calculated using various risk assessment models.(4) Throughout the family history ascertainment process and risk assessment discussion, the genetic counselor will assess the patient's own perception of his risk and how he appears to cope with and assimilate the information he is being provided.

Since genetic diseases are caused by an intrinsic error in the body that cannot be corrected, telling a patient that s/he has such a disease may be detrimental to his or her self-esteem.(11) Anxiety levels will directly affect how well the patient hears and understands the sometimes complex information being presented. In addition, patients often become emotional during the family history review when discussing either their current cancer diagnosis or that of a relative who is very sick or has died of cancer. Acknowledging and normalizing these psychological responses is crucial to the effectiveness of the genetic counseling session.

This patient's family is currently being evaluated using predisposition genetic testing for a mutation on the CDH1 gene that is responsible for hereditary diffuse gastric cancer. This cancer syndrome is inherited in an autosomal dominant manner.(12) If we locate a CDH1 mutation in the family, the patient will have a 50% risk of having inherited this mutation from his father. The results of molecular analysis are currently pending. The genetic counselor will be available to answer further questions regarding the genetic test results as well as to provide genetic counseling to any of the patient's at-risk relatives who may be at increased risk for gastric cancer.

Barriers to Incorporating Genetic Counseling

Before the potential for disease prevention as a result of new discoveries in genomics can be realized, a number of existing barriers must be overcome. The first is proper identification of patients with a potential genetic risk. Referral to the appropriate genetic services is contingent on the ability of the physician, and often the primary care physician, to effectively screen a patient's family and medical history for genetic susceptibility.

In his keynote address at the 8th Annual National Coalition for Health Profession Education in Genetics Conference, US Surgeon General Richard H. Carmona, MD, stated that a complete (3-generation) pedigree must be obtained for every child before birth.(13) He added that this family history will be the starting point for personalized, preventive medicine in the era of genomic medicine.

Inadequate family history

Despite Dr Carmona's recognition of the importance of documenting family history, studies suggest that family history information is not being used properly, if at all, as a screening tool. One study found that oncologists did not adequately document important family history information in almost one third of 362 patients and that 16% of those with a personal or family history suggestive of a hereditary cancer syndrome were not properly recorded as such.(14) A more recent study examining the use of a genetic family history as a risk assessment tool in an internal medicine clinic revealed that approximately 20% of 78 patients evaluated for genetic risk factors were not correctly noted to be in the appropriate genetic high-risk category.(15)

The knowledge gap

A second barrier to providing adequate genetic assessments for patients is a lack of physician interest and expertise. In a surveillance study of 1251 American physicians from 8 specialties, more than 89% said that physician guidelines for genetic testing for cancer susceptibility are needed, and only 29% felt competent enough to provide genetic counseling.(16) Among primary care physicians, only 40% said they regarded themselves as qualified to recommend genetic testing to their patients. Yet in another survey of 375 patients, 60% expressed interest in discussing cancer-related genetic counseling with their primary care physician.(17)

Studies report that physicians' time constraints and lack of interest, training, or understanding of medical genetics may prevent them from addressing genetic concerns with their patients.(18,19) An investigation into the use and interpretation of genetic testing for the hereditary colon cancer syndrome, familial adenomatous polyposis, found that testing was ordered erroneously for 20% of 177 patients and that only about 19% of patients received genetic counseling before the test.(18) In addition, the results were misinterpreted by physicians in about one third of cases.

Genetic Services in Primary Care

Primary care physicians who are not interested or lack the time to learn medical genetics can eliminate these barriers and meet the growing need and demand for genetic services by consulting with or referring patients to genetic counselors. Such counselors have specialized degrees in medical genetics and can support physician practices by incorporating the necessary family history screening information, interpreting the data, and discussing issues related to hereditary disease and suggesting possible genetic testing for a specific patient. Evidence indicates that incorporating a genetic counselor into patient care improves the detection of genetic risk factors.(20)

In 1975, the **American Society of Human Genetics** provided the formal definition of genetic counseling, calling it a communication process dealing with the problems associated with the occurrence or risk of a genetic disorder in a family.(21) Genetic counseling involves providing medical advice to a patient or a family in the attempt to help them better understand their disease and make appropriate treatment choices. This process helps patients and families:

1. Comprehend the medical facts about the diagnosis, its probable course, and the available treatment options.
2. Appreciate how heredity contributes to the disorder, and its risk of recurrence in specified family relatives.
3. Understand the alternatives for dealing with the risk of recurrence.
4. Choose a course of action that seems appropriate in view of the specific risk, family goals, and ethical and religious standards.
5. Make the best possible adjustment to the disorder and/or its risk of recurrence in an affected family member.

This definition, however, no longer covers the wide spectrum of disease and the specific counseling situations that have begun to emerge as a result of the Human Genome Project, including predisposition genetic testing, which genetic counselors are now trained to address.

After completing an accredited 2-year genetic counseling program, genetic counselors can become certified by completing the board competency examinations administered through the American Board of Medical Genetics or the American Board of Genetic Counseling. Some nurses also receive special training in medical genetics, but many do not obtain board certification, given that eligibility to sit for the boards requires attendance at an accredited training program in genetic counseling.(22)

Genetic counselors often work in conjunction with clinical geneticists, who are physicians that have been trained to diagnose and treat genetic diseases as well as provide genetic counseling. In an ideal model of patient care for genetic diseases, the primary care provider would lead the team and coordinate the patient's overall health care. The clinical geneticist would be asked to obtain a diagnosis and initiate treatment. The genetic counselor would educate and counsel the patient before and after genetic testing and address psychosocial and/or psychological concerns arising from the diagnosis.

Often, laboratory-trained geneticists, such as cytogeneticists, molecular geneticists, and/or biochemical geneticists, are involved in interpreting the patient's genetic test results. Other specialists may also be involved in patient management. Additional health care professionals, such as nurses, social workers, and

nutritionists, are important contributors to the management of a patient with a genetic disease.(23)

Nondirective Counseling

Consistent with genetic counselors' practice of "nondirective counseling," the decision about whether to undergo genetic testing, if applicable, is one that the patient is empowered to make autonomously. Issues reviewed with the patient in the informed consent process include but are not limited to²⁴:

1. A discussion of the purpose of the test (eg, research versus clinical).
2. The cost of the test, its turnaround time, and how the results will be disclosed to the patient.
3. Predictive value of a positive, negative, or indeterminate result, and corresponding cancer risk information.
4. Options for cancer risk management if the result is positive, negative, or indeterminate.
5. The potential psychological implications of testing.
6. Discussion of concerns, such as the potential for insurance or employment discrimination.
7. A review of alternatives to genetic testing, including delaying the decision to pursue it.

If genetic testing is ordered, the patient will be scheduled for a follow-up visit with the genetic counselor, during which the test results will be disclosed and the medical recommendations derived from the family history, and/or the test results will be summarized. Results of any genetic test are also distributed to the primary care physician and other physicians participating in the patient's care, such as an oncologist or surgeon. The patient is then referred back to the primary care physician for a discussion of future management for either treatment of disease, if the patient already has cancer, or for prevention, if the patient is healthy but carries a hereditary cancer susceptibility gene.

The entire process of genetic counseling for hereditary cancer syndromes may include 2 or more sessions, each lasting between 60 and 90 minutes.

When to Refer

Table 2 lists common indications for referral to genetic counseling in the primary care setting. Referrals for hereditary cancer syndromes provide only one example of how a primary care physician can benefit from teaming up with genetic counselors. Other examples of when to refer patients include the following 2 cases: a 40-year-old man with 2 children, aged 15 and 12 years, who has polycystic kidney disease and wants to know about his children's chances of also being affected; another is a 34-year-old man whose father was diagnosed with Wilson's disease and is concerned that he, too, could be affected. Genetic counselors could help the primary care physician in assessing risk and providing a potential course of action.

Conclusion

Genetic counselors can team up with primary care physicians to establish referral protocols appropriate for additional genetic evaluation, review the family history of referred patients, provide risk assessment based on a family history, and, when appropriate, discuss genetic testing options and follow-up management. Genetic counselors enhance the quality of patient care and serve as advocates and educators for patients as well as educators for the medical and nonmedical communities.

Before the benefits of using the expertise of genetic counseling can be fully realized, primary care physicians will require more education about the value of this approach in patient care. Genomics is no longer an esoteric academic specialty; it is now critical to the delivery of effective health care and preventive medicine for all patients.(10)

Self-assessment test

1. Which of the following statements about genetic counseling is NOT true?
 - A. A thorough family history is crucial to genetic risk assessment
 - B. Most physicians today have adequate training in genetics

- C. Incorporating genetic counseling into patient care improves detection of genetic risk factors
- D. Genetic counselors can be part of a team led by the primary care physician
2. Which of the following situations is NOT an indication for genetic counseling for hereditary breast cancer?
- A. Breast cancer occurrence in a 55-year-old woman
- B. Breast cancer occurrence in a 70-year-old man
- C. Breast cancer in a woman with a history of ovarian cancer
- D. Breast cancer and oral papillomatosis
3. Genetic counseling for the possibility of hereditary colon cancer is indicated in all these patients, except:
- A. A 35-year-old woman with 1 adenoma
- B. A 60-year-old man with 8 adenomas
- C. A 45-year-old woman with colorectal cancer
- D. A 65-year-old man with 2 family members with sebaceous carcinomas
4. All these are indications for referral to genetic counseling, except:
- A. Family history of cystic fibrosis
- B. Diagnosis of thalassemia
- C. Family history of Wilson's disease
- D. Family history of hypopituitarism
5. Which of these is NOT an indication for preconception or pregnancy genetic counseling?
- A. Abnormal fetal ultrasound findings
- B. Evaluation of a birth family before adoption
- C. Pregnant woman who will be 32 years old at time of delivery
- D. History of 4 spontaneous abortions

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