Outpatient Genetic Risk Assessment in Women With Breast Cancer: One Center’s Experience

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The Human Genome Project was completed on April 14, 2003, and resulted in a greater understanding of the underlying genetic etiology of cancer. Although not all cancers are caused by inherited genetic mutations, all do require the accumulation of a series of acquired somatic mutations that eventually render healthy cells malignant. Because of a rapidly developing knowledge base in genetics, healthcare professionals are expanding their practice by offering more comprehensive options for cancer risk assessment and screening. Cancer genetics, a legitimate oncology nursing subspecialty, has implications for nurses in all practice settings and should be incorporated into general and advanced nursing practice (Greco, 2003).

The Oncology Nursing Society (ONS), the American Society of Clinical Oncology (ASCO), and the National Cancer Institute have developed guidelines for incorporating genetic information into clinical practice (ASCO, 1997; Barse, 2003; Garber et al., 1997; Greco, 2003; Stopfer, 2000). Research groups also have recognized the importance of developing evaluation programs to address the complex issues associated with genetic testing for cancer risk (Barse; Calzone, Stopfer, Blackwood, & Weber, 1997; Greco) and the implications for healthcare practitioners (Barse; Calzone, 1997; Greco).

Cancer risk assessment and education, facilitation of genetic testing, pre- and post-test counseling with individualized cancer risk management options, and supportive care are integral to the provision of cancer genetics services to patients (Greco, 2003). Healthcare professionals should be familiar with the complex issues surrounding genetic testing and counseling and have the specialized training to provide these services (Hutson, 2003).

Methods

Developing the Research Question

In January 2002, the Quality Initiative (QI) Committee at the authors’ institution decided to address the issue of appropriate recognition of genetic risk information in clinical practice. Their goal was to ascertain whether oncologists documented the family history of patients and accurately interpreted cancer risk as well as to determine the rate of referral to genetic counselors. A literature review revealed little regarding whether healthcare practitioners use guidelines on incorporating genetic screening into oncology practice. An algorithm was developed as a guide to improve the process of evaluation and referral for genetic risk assessment.

Sample Selection

The authors’ institution is a community-based cancer care organization with six sites.

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Although all patients should be assessed for the potential of a genetic mutation, patients with breast cancer were chosen because of the large patient population. The incidence of genetic mutation is associated with a younger age at diagnosis; therefore, the sample only included women who were diagnosed before age 65. Patients currently undergoing chemotherapy were eliminated from the sample; they were unlikely to be referred during treatment as a result of factors that included emotional distress and side effects. Therefore, women treated at the clinic from February 1–March 31, 2002 (N = 1,100), who were younger than age 65 and had a diagnosis of breast cancer, were identified as potential subjects. From this population, the final audit sample (n = 193) was randomly selected to allow at least 36 patients per clinic site and represent each oncologist’s practice.

Chart Audit

The chart audit was completed by five advanced nurse practitioners and one oncology nurse from the genetics committee. Prior to the audit, four nurse practitioners attended a Cancer Risk Educator Workshop that was offered to physicians and nurses in the community. An audit tool was developed by the QI Committee with the help of the cancer genetics counselor and an oncologist. Elements to be assessed in the chart audit included a genetic risk assessment using a family history pedigree and risk reduction strategies. The completed pedigree included information regarding all documented types of cancer found in family members, the age at diagnosis, and whether those family members died of cancer. Documented risk reduction strategies included more frequent screenings, chemophylaxis, or referral to a cancer genetics counselor. The charts were scored based on the completeness of the documented family history and recorded risk reduction strategies (see Table 1). Inter-rater reliability was not assessed because each member of the audit team was present and participated in discussions during the development of the audit tools. Scenarios were discussed during instrument development, and each of the auditors verbalized understanding of the scoring criteria.

Results

Demographics

Within the sample population, the average age at diagnosis was 50.25 years (SD = 6.80). Most patients (64.25%) were 45–59 years old (see Table 2) and Caucasian (59%). Few non-Caucasians were randomly selected to be audited, which may reflect the population at each of the centers.

Family History of Cancer and Risk Management

Patients received a score of zero, one, or two based on the amount of family history information documented in their charts. Eleven percent of those audited received a score of zero, indicating that no family history information was recorded. Most patients received a score of one or two, indicating that their family history had been documented. Omission of family history occurred because the information had not been received from the hospital where the patient was treated or the physician recently moved from another practice and failed to record histories for patients he or she had treated previously. Of the 193 charts reviewed, 88% had some documented family history and 78% had some intervention for risk management documented (e.g., routine cancer prevention screening). Less than 10% of patients were considered for genetic counseling referral.

Discussion

The current understanding of the nature of hereditary cancer and the availability of genetic testing for cancer predisposition show that many patients and their families may benefit from careful assessment of their hereditary cancer risk. Detailed questions regarding family medical history should be a standard component of the initial intake for any patient entering the healthcare system. A family history questionnaire can be used as a screening tool to identify those who could benefit from detailed evaluation of familial cancer susceptibility (ASCO, 2004). This chart audit suggests that, in some settings, providers do obtain some family history, but often not to three generations. Because of the lack of documented three-generation family history, patients who have a genetic predisposition cannot be assessed accurately; therefore, management of cancer risk and referral to a genetic risk counselor may not occur.

Strong hereditary components exist in roughly 5%-10% of all patients with breast cancer (ASCO, 2004; Lashley, 1998). The 7.25% overall rate of referral to genetic counseling in this audit may indicate that the physicians had identified individuals with potentially hereditary breast cancer who had the greatest need for genetic testing services and may reflect an appropriate use of genetic counseling services among the physicians. Alternatively, approximately 15%-20% of breast cancer cases are associated with some family history of the disease but have no evidence of autosom-
mal dominant transmission. The “familial clusters” of breast cancer are thought to result from weaker genetic predisposition as well as environmental factors such as diet and lifestyle. As a result, at a 7.25% referral rate, some women may not be assessed accurately for hereditary risk (ASCO, 2004).

Moreover, nearly half of the family medical histories recorded in the patient charts did not include three generations of family history. To fully assess most families, three generations on both sides of the proband’s family should be recorded (Bennett et al., 1995; Lashley, 1998), which, ideally, includes documenting the health status of all individuals (i.e., those with and without cancer diagnoses) and defining the pathologic cancer types and age of onset of all cancer cases.

Risk reduction strategies should be employed for all patients diagnosed with cancer. Many patients are not educated about screening recommendations such as colonoscopy, mammography, breast self-examination, digital rectal examinations, or Pap tests. More specific risk reduction strategies need to be recommended when patients are at higher risk for developing cancer.

**Recommendations**

Based on these audit findings, a number of new projects were initiated to improve access to genetic counseling for patients. An algorithm for patient referral for genetic counseling, based on ASCO (1997) guidelines, was developed (see Figure 1).

A self-administered family history questionnaire (see Figure 2) to address the issue of incomplete collection of family history data was incorporated into this center’s practice. The following cancers are listed on the questionnaire: breast, endometrial, ovarian, lung, colon, prostate, pancreatic,
and other. Patients provide information about maternal, paternal, and first-, second-, and third-degree relatives. Once completed, the questionnaire is reviewed by the physician and maintained in the patient’s permanent medical record. At the end of therapy, an advanced practice nurse reviews the family history and initiates genetic counseling when appropriate and according to ASCO (1997) guidelines.

Another outcome of the audit was the recognition of the need for further education in genetics for physicians and nurses. Healthcare professionals may lack the most recent knowledge necessary to help individuals make informed decisions about genetic counseling and testing. Also, professionals may not fully realize the impact of genetics on patient decisions, such as the option of prophylactic surgery at the time of diagnosis for women who carry a genetic mutation (Van Riper & McKinnon, 2004).

Many opportunities for nursing education in genetics exist, such as the comprehensive course conducted by ONS in conjunction with the National Cancer Institute. This course covers the basics of Mendelian genetics, the molecular biology of cancer, and the role of genetic testing for risk assessment among patients with cancer and their families. Six nurse practitioners from the authors’ institution attended this course, and all successfully demonstrated 24 of the 43 competencies established by the National Coalition for Health Professional Education in Genetics. Additionally, they have conducted in-service educational events to disseminate this information. Physicians were educated about the results of the project and its implications for their practices. The physicians recognize nursing as a resource for genetic risk assessment methods.

**Summary**

These findings support the need to improve genetic risk assessment. Improvements were made to the family history collection tool, and a clinical algorithm was developed to aid in the management and referral of patients at high risk for developing breast cancer. The advance practice nurses and nursing staff underwent further training in genetics. The algorithm will be updated regularly to incorporate the latest ASCO policy regularly and used to make the appropriate referrals based on the degree of risk for the patient or the patient’s desire for referral.

Preliminary data suggest that referrals have increased by as much as 400% since the implementation of the new procedures. Future chart audits will evaluate the impact of the algorithm and improved family history assessments and will include the number of patients whose referral for genetic counseling was appropriate for their genetic risk. The expected outcomes include improved recognition of women who have a possible genetic predisposition to cancer and appropriate referrals to a cancer genetics counselor. Following evaluation of this program, it may be expanded to include patients with colon cancer.

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**References**


Rapid Recap

Outpatient Genetic Risk Assessment in Women With Breast Cancer: One Center’s Experience

- A recorded family history spanning three generations enables nurses to target patients who are at high risk for developing cancer and have the greatest need for risk management programs.
- Assessment tools for gathering information about women with breast cancer were compared to test for reliability and recognition of familial cases, an important indicator of genetic risk.
- A risk management tool and pedigree template were developed to examine the completeness of the recorded family cancer history.
- An algorithm was developed as a guide to improve the process of evaluation and referral for genetic risk assessment.


