

Inheriting Breast Cancer:

Genetic counseling for the BRCA 1 and BRCA 2 genes

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Introduction

Breast cancer is the second leading cause of cancer-related death in women in the United States, and it will take the lives of approximately 40,460 women in 2007 (American Cancer Society, 2007). Positive family history is known to increase risks of breast cancer, but only 5% to 10% of cancers are genetically inherited (MacDonald, 2002). The most common causes of this hereditary breast cancer comes from mutations in the BRCA1 and BRCA2 genes which increase a woman's risk of breast cancer to as high as 85% (MacDonald, 2002). In 1996, the availability of commercial testing to screen for these gene mutations brought new risk management options to the general population and a new set of ethical issues to ponder (MacDonald, 2002). As medical professionals, it is important to understand the implications that this kind of genetic testing has on the client and on health care. The purpose of this paper is to provide the reader with information about the implications of having an increased risk for breast cancer, the risk management options that genetic counseling provides, and the professional's role in genetic counseling.

Review of Literature

In order to understand the importance of genetic counseling for BRCA1 and BRCA2, we must look at how having a family history of breast cancer affects the individual. Women who have a family history of breast cancer often overestimate their own risk of developing breast cancer (Matloff, Moyer, Shannon, Niendorf, & Col, 2006). These high risk individuals live in a state of "chronic risk" that can have many "physical, social, cultural, financial, and medical meanings and can affect feelings of self-worth as well as interpersonal relationships" similar to individuals suffering from an actual chronic illness (Kenen, Arden-Jones, & Eeles, 2003, p. 316).

Most of these women with a strong family history of breast cancer have experienced the agonizing effects of cancer on a loved one, so it is easy to see how these life experiences would have a profound effect on their health risk perceptions and behaviors (Matloff et al., 2006). Genetic counseling provides a technological advantage that can allow for increased screening and preventative interventions for those identified as carriers of a BRCA1 or BRCA2 mutation gene, as well as freeing those identified as non-carriers from the stress of intensive medical screening or prophylactic surgery (Claes et al., 2005). Once the carrier status has been established, decisions must be made about risk management options which are discussed as part of the genetic cancer risk assessment (MacDonald, 2002).

Options for risk management of breast cancer include screening, chemoprevention, and prophylactic surgery (MacDonald, 2002). Screening methods used for early detection of breast cancer include mammograms annually after age 40, annual clinical breast exams, and monthly self breast exams. For individuals identified as having a high risk for breast cancer, these screening methods are started at younger ages and may include an ultrasound or MRI with the annual mammogram (American Cancer Society, 2007). Chemopreventive agents such as tamoxifen are now available to reduce the risk of breast cancer by as much as 50% in healthy high risk women. However, this drug carries risks of its own ranging from vaginal discharge and hot flashes to side effects as severe as increased risk for endometrial cancer and thromboembolic events (Matloff et al., 2006). In one study, over 90% of the women who qualified for chemopreventative medication declined with adverse reactions as the most commonly cited reason (Matloff et al., 2006). Prophylactic surgery is also an option including bilateral mastectomies and prophylactic oophorectomies to minimize the risk for ovarian cancer that is also associated with the BRCA1 gene mutation; although it is important to note that these

surgical procedures may not eliminate the risk in these genetically predisposed women (MacDonald, 2002). Clearly these are not easy choices. This puts the healthcare professional in a unique role in this decision-making process.

Genetic counseling has traditionally consisted of a nondirective approach wherein the counselor provides all the information for the client to interpret and use in their “autonomous decision making” (Evans, Bergum, Bamforth, & MacPhail, 2004, p. 461). Despite the healthcare provider’s obligation to respect the client’s autonomy, studies show that many women desire recommendations from their physician in this delicate matter. Some women want to be given a specific course of action while others want to be presented with all the options (Geller, Strauss, Bernhardt, & Holtzman, 1997). These findings indicate the need for an adaptation of this traditional nondirective approach to a holistic approach in which there is an active dialogue between the client and the healthcare provider thereby enhancing both autonomy and beneficence (Evans et al., 2004). This approach requires healthcare providers to “disclose their own motivations, beliefs, and values” and ask their clients questions that yield insight into their health beliefs, practices, and experiences (Geller et al., 1997, p.31). This does not mean that the provider should exercise paternalism or try to influence the client’s decision, but rather strive to see the client as a unique and whole person who is not just another client with the BRCA mutation gene (Evans et al., 2004).

Genetic counseling in this particularly vulnerable population involves the timing of the genetic testing and sensitivity when providing information and counseling. Women who had been diagnosed with breast cancer indicated that they did not want to handle both their diagnosis and genetic testing at the same time because they would feel overwhelmed by the information and therefore be more likely to rush towards a decision that they are not yet ready to make and

may later regret (Arden-Jones, Kenen, & Eeles, 2005). Occasionally women with a family history of breast cancer choose to have prophylactic mastectomies regardless of the outcome of their genetic testing (Geller et al., 1997). Studies show that there is “no perfect time” to offer genetic testing, and that the “right time” only occurs when the client is ready for the information (Arden-Jones et al., 2005, p. 275).

Another issue debated by both clients and professionals is who should handle the genetic counseling. Women as a whole seem to feel that it is best to have the genetics department handle the counseling so they would know that they are dealing with an expert who can offer plenty of time to ensure that the client is comfortable, accurately informed, and not rushed (Arden-Jones et al., 2005). In contrast, medical professionals generally agreed that this counseling should be handled by the breast surgeons or the oncologists. Surgeons feel that there was no need for women to see the genetics department when they already are interacting with a surgeon who was quite familiar with the genetic aspect of this disease (Arden-Jones et al., 2005). Other healthcare professionals, including breast cancer nurses, did acknowledge the time constraints at busy clinics. They felt that it was more important to make sure that the client knew the testing was available and then provide answers to any questions that the client might have (Arden-Jones et al., 2005). It is clear from these differing opinions that the delicate matter of genetic counseling must be handled on an individual basis and not generalized.

Conclusion

Nurses must take into consideration that clients with a family history of breast cancer often overestimate their own risks leading to a “chronic risk” lifestyle that can affect many aspects of their lives (Kenen et. al., 2003, p. 316). For these women, the BRCA1 and BRCA2 genetic testing offer a sense of “certainty” about their high risk status and provides them with the

information necessary to make informed decisions about their risk management options (Claes et al., 2005, p.102). These options, which include screening, chemoprevention, and prophylactic surgery, are not without their own risks and therefore require careful thought and consideration. The professional's role in genetic counseling is a very valuable one involving a balance between respecting the client's autonomy and maintaining beneficence. Generating a counselor-client relationship that fosters an open dialogue and provides the client with the support necessary to deal with this life-altering issue is vital. The timing of the genetic testing and the type of professional that provides the counseling are two ways in which this process can be tailored to the client's individual needs for improved comfort and effectiveness.

The prevalence of breast cancer in our society today makes nurses responsible for understanding these issues and exploring our own opinions. Regardless of their specialty, nurses will encounter a woman with a family history of breast cancer and have the opportunity to meet that woman's needs by having adequate, accurate, and thorough information to assist her in the decision-making processes (MacDonald, 2002). As a future nurse and a woman with a family history of breast cancer, this issue holds both professional and personal relevance and I value the insight gained from reviewing the literature and writing this paper. As nurses we pride ourselves on being patient advocates, and so we must assist these high risk women by making sure they know genetic testing is available and the various risk management options as well as ensuring that their needs are met throughout the genetic counseling process. Ultimately this makes us better nurses and increases the overall health of our clients and the population as a whole.

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