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Women with Genetic Mutation Conferring High-Risk for Breast Cancer

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Abstract

Estimation of breast cancer risk for a woman who does not have a personal history of invasive breast cancer or ductal carcinoma in situ begins with an initial assessment of familial/genetic factors associated with increased breast cancer risk for the purpose of determining whether more extensive genetic risk assessment and counselling should be undertaken. The first step in this primary assessment is a broad and flexible evaluation of the personal and family history of the individual, primarily with respect to breast and/or ovarian cancer/fallopian tube or primary peritoneal cancer.

Keywords: Genetic component; General population; Inheritance patterns; Sporadic cancer; Penetrance genes; Weight gain

Introduction

Genetic predispositions conferring a high risk for breast cancer include hereditary breast and ovarian cancer, Li-Fraumeni syndrome, Peutz-Jeghers syndrome, Cowden syndrome and hereditary diffuse gastric cancer. If the individual has a known genetic predisposition for breast cancer such as mutations in BRCA1/2, TP53, PTEN, or other genes associated with breast cancer risk, that individual must be counselled about risk reduction options. If the familial/genetic factors are not known, a thorough evaluation must be performed [1]. The magnitude of the risk increases with the number of affected relatives in the family, the closeness of the relationship, and the age at which the affected relative was diagnosed. The younger the age at diagnosis of the first- or second-degree relative, the more likely it is that a genetic component is present [2]. The maternal and paternal sides of the family should be considered independently for familial patterns of cancer. Hereditary cancers are often characterized by gene mutations associated with a high probability of cancer development, vertical transmission through either mother or father, and an association with other types of tumors. They often have an early age of onset and exhibit an autosomal-dominant inheritance pattern [3]. Familial cancers share some but not all features of hereditary cancers. For example, although familial breast cancers occur in a given family more frequently than in the general population, they generally do not exhibit the inheritance patterns or age of onset consistent with hereditary cancers. Familial cancers may be associated with chance clustering of sporadic cancer cases within families, genetic variation in lower penetrance genes, a shared environment, or combinations of these factors. If an individual or a close family member of that individual meets one or more of the criteria listed in the NCCN Guidelines for Breast Cancer Risk Reduction under Familial Risk Assessment, that individual may be at increased risk for familial/hereditary breast cancer, and referral for formal genetic assessment/counselling is recommended [4]. A cancer genetics professional should be involved in determining whether the individual has a lifetime risk for breast cancer greater than 20% based on models dependent on family history. The Claus tables may be useful in providing breast cancer risk estimates for white women with no known cancer-associated gene mutation but who have one or two first- or second-degree female relatives with breast cancer and ovarian cancer. Breast and Ovarian Analysis of Disease Incidence and Carrier Estimation Algorithm are more commonly used to estimate the risk of a mutation. Strong genetic association between breast and ovarian cancer has been demonstrated in some families by linkage analyses [5].

Methodology

Based on a risk assessment using one or more of these models, women with a BRCA1/2, TP53, or PTEN gene mutation, or a pedigree strongly suggestive of genetic predisposition to breast cancer, may be identified. The NCCN Guidelines for Genetic/Familial High-Risk Assessment, Breast and Ovarian describe management strategies for women with a known or suspected BRCA1/2, TP53, or PTEN mutation or a pedigree strongly suggestive of genetic predisposition to breast cancer. Other Elements of Risk For women not considered to be at risk for familial/hereditary breast cancer, an evaluation of other elements of risk that contribute to increased breast cancer risk is recommended as shown in (Figure 1). These include demographic factors such as

Figure 1: Treatment advance for breast cancer.

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female gender, age, and ethnicity/race. There is an increased incidence of BRCA1/2 mutation reported in women of Ashkenazi Jewish decent. Reproductive history is another factor to consider [6]. Risk factors linked to reproductive history include null parity, prolonged interval between menarche and age at first live birth, onset of menarche at a younger age, or onset of menopause at older age. Body mass index is an independent risk factor for breast cancer, especially in Caucasian women. Several studies have established the association between high BMI and adult weight gain and increased risk for breast cancer in postmenopausal women. This increase in risk has been attributed to increase in circulating endogenous estrogen levels from fat tissue. In addition, the association between BMI and risk for postmenopausal breast cancer is stronger for hormone-positive tumors. A meta-analysis of more than 1000 epidemiologic studies looked at cancer risk with excess body fat [7]. Women with higher BMI experienced an increased risk of postmenopausal breast cancer. Lifestyle factors such as current or prior hormone therapy, alcohol consumption, and, to a lesser extent, smoking also contribute to the risk of developing breast cancer as shown in (Figure 2). The risk for breast cancer associated with FEA is similar to that of benign proliferative disease without atypia. The data are not as strong with respect to the degree of risk or the benefits of risk-reduction therapy in this population. Proliferative lesions with atypia include atypical ductal hyperplasia, atypical lobular hyperplasia, and LCIS. These lesions are associated with an increased risk of developing breast cancer. Women with LCIS are at substantially increased risk for breast cancer. In a population-based study of 19,462 women diagnosed with LCIS from the SEER database between 1983 and 2014 in which the cumulative incidences of subsequent breast malignancy were 11.3% and 19.8% at 10 and 20 years, respectively. At a median follow-up of 8.1 years, primary breast cancer was diagnosed in 9.4% of the cohort [8]. Other factors to consider are number of breast biopsies, done with the intent to diagnose cancer. Change in breast density has been suggested as a risk factor for breast cancer. Dense breast tissue as measured by mammography is increasingly recognized as an important risk factor for breast cancer. For example, a report of a large case-cohort study of women 35 years and older with no history of breast cancer who underwent mammographic screening, first at baseline and then at an average of 6 years later, suggested that longitudinal changes in breast density are associated with changes in breast cancer risk. There are many elements that may reduce the risk of cancer. Breast feeding has been shown to have a protective effect in many studies [9].

Figure 2: Higher BMI experience an increased risk of breast cancer.

Discussion

An analysis of 47 epidemiologic studies estimated that for every 12 months of breastfeeding, RR for breast cancer decreases by 4.3%.82 exercises has also been shown to reduce the risk of breast cancer, especially in post-menopausal women. A most recent review of epidemiologic studies estimated that risk of breast cancer was reduced among women who were most physically active compared with those who were least active [10]. Oophorectomy before age 45 years and risk-reduction therapy have a protective effect. A large prospective study examined associations of hysterectomy with bilateral salpingoophorectomy and simple hysterectomy in 66,802 postmenopausal women from the Cancer Prevention Study-II Nutrition Cohort. The results showed that hysterectomy with BSO performed at any age, compared with no hysterectomy, is associated with a 10% reduction in all cancers. Cancer The modified Gail model is a computer-based, multivariate, logistic regression model that uses age, race, age at menarche, age at first live birth or null parity, number of first-degree relatives with breast cancer, number of previous breast biopsies, and histology of the breast biopsies to produce actuarial estimates of future breast cancer risk. Women ≥35 years of age should have their risk for breast cancer estimated according to the modified Gail model [11]. The Gail model is not an appropriate breast cancer risk assessment tool for women with a BRCA1/2, TP53, or PTEN mutation; a strong family history of breast cancer; women who received thoracic radiation to treat Hodgkin's disease, or those with LCIS. While the Gail model can overestimate the risk for some women, in some others, notably women with AH, it can underestimate their risk making them appear to be ineligible for risk-reduction therapy [12]. The Gail model does not apply to women with FEA. The risk threshold required to consider the use of risk-reduction strategies must depend on an evaluation of the efficacy, morbidity, and expense of the proposed intervention. As a reasonable discriminating threshold, the NCCN Breast Cancer Risk Reduction Panel has adopted the 1.7% or greater 5-year actuarial breast cancer risk as defined by the modified Gail model, which was used to identify women eligible for the NSABP Breast Cancer Prevention Trial and the Study of Tamoxifen and Raloxifene trial [13]. The Tyrer-Cuzick model, in addition to considering a woman's risk of a BRCA mutation, also estimates her risk of developing breast cancer using not only family history but also epidemiologic variables including a personal history of AH or LCIS [14]. Women with AH or a history of LCIS are also at substantially increased risk for invasive breast cancer in both the affected and contralateral breast [15]. In an analysis of the Mayo Clinic cohort of more than 300 women with AH, the Gail model underestimated breast cancer risk for women with AH, whereas the Tyrer-Cuzick model overestimated this risk. Breast density is not included in any of the commonly used risk assessment models/tools.

Conclusion

The innate immune cells belonging to myeloid lineage composed of TAMs and immature myeloid cells are found to be involved intrinsically. These cells produce various chemokines, cytokines, proteases and several growth factors, which may promote tumour growth; and mediate local or systemic immune-suppression by inducing angiogenesis and tissue remodelling.

Acknowledgement

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Conflict of Interest

None

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