BRCA1 Gene Mutation and Breast Cancer Literature Review

By: Megan Wells
April 22, 2016

Abstract: Research about breast cancer and the BRCA1 gene mutation was completed using a systematic review of many different resources. Through researching these topics separately and then in relation to one another, the researcher was able to gain insightful knowledge into the bidirectional relationship breast cancer and the BRCA1 gene have with one another. Through examining and analyzing multiple different peer-reviewed sources, a variety of research covering different facets of breast cancer and the BRCA1 gene was obtained. This multidisciplinary research allowed for a more well rounded understanding of the topics.

Objectives:

To review the research completed on the relationship between the BRCA1 gene mutation and breast cancer onset.

This assignment was used to fulfill a BBH411W requirement of a short paper assignment acting as a Literature Review.

*Disclaimer: The purpose of the writing is to fulfill course requirements for BBH 411W and to stand as a personal writing sample, but the findings should not be treated as generalizable research.
The Disease:

Breast cancer represents the most commonly diagnosed form of cancer in women, affecting about one in eight women in the United States. It also represents the second highest cause of death from cancer with about 40,000 mortalities per year in the United States (Ahmad, 2013). Since breast cancer affects about 249,260 people per year, it proves to be extremely valuable to continue researching this disease in order to fully understand the development and epidemiology of breast cancer (Siegal et al., 2016). Breast cancer detection is usually by self-examination at home and then by a mammography in a doctors’ office. Studies show that the populations who utilize the mammography have a 31% decreased breast cancer mortality rate (Tabar, 1985). Therefore the mammography is a tool that can be effectively utilized to detect breast cancer earlier and reduce the mortalities resulting from breast cancer.

In order to fully understand the disease, one must first address the origin, and why breast cancer affects specific individuals compared to others. Although it is not possible to pinpoint the exact cause of breast cancer onset, the disease develops as a result of a gene and environment interaction. Damaged cell DNA can frequently lead to genetic mutations, which then causes breast cancer to develop. But this damaged cell DNA can be a result of a genetic or environmental occurrence in an individual’s life (Risk Factors: The National Breast Cancer Foundation). There are also many different risk factors that increase the likelihood of breast cancer onset including sex, age, race, and family history. Sex is an important risk factor because as a woman, an individual is 100 times more likely to occur than for a man. Age also proves to be important because as an individual gets every ten years older, their risk of getting breast
cancer increases by 50% (McPherson, 2000). Being Caucasian also represents an extremely important risk factor since Caucasian women represent a group with higher incidence rates when compared to other races. Family history represents an extremely important risk factor for breast cancer because since the cancer can occur from a genetic mutation, this mutation can be passed down from generation to generation. Therefore, if an individual in someone’s immediate family suffered from breast cancer, she has a higher rate of also getting breast cancer. Although, if the family member who had breast cancer is not related by a first-degree relationship, the risk of inheriting breast cancer from them significantly decreases (Anderson, 1974). Anderson (1974) also concluded through his research that a mother had a 30% chance of passing down a breast cancer gene to her daughters. Her daughters then also had a 30% higher chance of breast cancer onset before the age of 30. This concludes that breast cancer related to a genetic heritability often occurs at a younger age compared to someone who gets breast cancer from a more environmental cause. The heritable aspect of breast cancer also accounts for about 5 to 10% of all cases (McPherson, 2000). There are also external factors that contribute to breast cancer onset as well including hormone imbalance, smoking, weight, alcohol intake, and many others (Anderson & Matsuno, 2006).

**BRCA1 Gene**

The BRCA1 gene, an abbreviated name for the Breast Cancer 1 gene, is frequently linked with breast and ovarian cancer in women. Therefore, being a carrier of the BRCA1 gene highly increases one’s risk for breast cancer onset (Chen & Parmigiani, 2007). Chen and Parmigiani (2007) completed a meta-analysis that analyzed how significant being a carrier of the BRCA1 gene was in relation to early onset breast cancer incidence. They found that individuals who
carried the BRCA1 gene reported p-vales of 0.001-0.045. This data supported the hypothesis that having this gene significant increases an individual’s risk of early onset breast cancer (Chen & Parmigiani, 2007). This statistic is due to the function of the BRCA1 gene and how the gene’s mutation can lead to breast cancer.

The BRCA1 gene is responsible for producing a tumor suppressing protein that aids in repairing damaged cell DNA. Therefore it is extremely problematic when a mutation in this gene occurs, often leading to the mutated cell to rapidly divide and multiply. Also the longer cell DNA is damaged and not repaired, the more likely it will be that it may mutate and develop into a certain type of cancer (Miki et al., 1994). As mentioned before, these mutations can then be passed down to children and lead to future breast cancer onset. When breast cancer occurs from a BRCA1 gene mutation, it is mostly seen in individuals below the age of 35 (Anglian Breast Cancer Study Group, 2000).
References


