BRCA for Beginners

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Introduction

The BRCA1 and 2 gene mutations aren't something that just happens, they often stem from family inheritance and are passed down through several generations of ancestors. Inherited mutations in BRCA1 and BRCA2 predispose to high risks of breast and ovarian cancer.[5] Its important to note that not all breast and ovarian cancer patients have that gene mutation. Most breast and ovarian cancers are sporadic (that is, not inherited), but some are the result of inherited predisposition, principally due to mutations in the tumor suppressor genes BRCA1 and BRCA2. [6] This is where the need for genetic testing can go a very long way. A person with a BRCA gene mutation should know about various methods that promote early detection and decrease the risk of developing cancer, including increased surveillance, chemoprevention, and pro-phylactic surgery.[8] Although the BRCA genes themselves appear unconnected to common, nonhereditary cancers, emerging evidence suggests that defects in other parts of the BRCA pathway might be critical not only in driving breast cancer but other cancers as well. [4] The purpose of this poster is to increase awareness of the mutations that go along with the BRCA genes and the risk that comes with. As well as bring about an understanding of what testing results mean, and the different screening techniques put in place to help prevent escalation of these mutations.

Positive Result: this result indicates that a person has inherited a harmful variant of BRCA1 or BRCA2 and has an increased risk of developing cancers
Negative Result: this result indicates a person did not inherit the harmful variant of BRCA 1 or 2.
Variant Of Uncertain Significance Result: this result is the closest to being undecided. It's unknown as to whether a specific gene is harmful.

Table 1: Each test result and a brief explanation of its meaning. [1]

Studies

- BRCA1 and BRCA2 are genes that produce proteins that help repair damaged DNA. These genes are called tumor suppressor genes because when they have certain changes, called harmful (or pathogenic) variants (or mutations), cancer can develop. [1]
- The incidence of *BRCA1* or *BRCA2* mutations within the general population is infrequent and only found in 1 out of every 300 to 800 people. Certain populations exhibit a higher likelihood of harboring genetic mutation than the general population. These include Ashkenazi Jewish patients, male patients who develop breast cancer, and patients younger than 30 years old who develop breast cancer. [3] (Figure 1)
- Testing for inherited BRCA1 and BRCA2 variants is usually done through a blood or saliva sample. Unless an inherited risk is known this is not recommended for the public. [1]
- A positive BRCA mutation indicates a higher likelihood of developing cancer but does not make or confirm the diagnosis of cancer. Subsequently, a negative *BRCA* test does not eliminate the risk of developing breast cancer from sporadic or other genetic causes. [3] (Table 1)
- Some people with a positive result might choose to take part in more frequent breast screenings at an earlier stage of life. (Table 2)

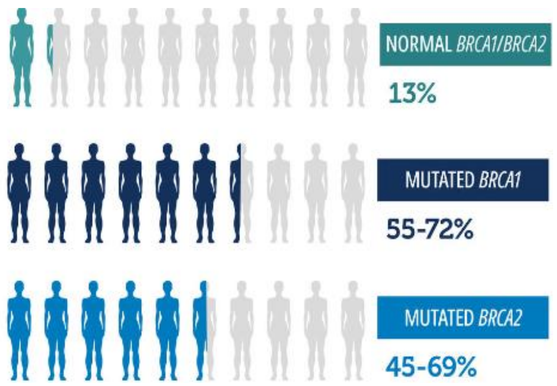

Figure 1: Percentages from a wide range of studies showing cancer risk following women up to 70-80 years old [2]

Increased surveillance/Screening: this includes... ▪ Self-examination, clinical breast exams, etc. ▪ Breast ultrasound ▪ Breast MRI ▪ Mammogram
Risk Reducing Surgery: this surgery removes as much of the "at risk" areas of the breast as possible, in hopes of reducing this risk of breast cancer as much as possible.
Chemoprevention: is administering or taking a given medication to lower the risk of developing, in this case, cancer.

Table 2: Each of the more common screening and prevention options. [1]

Discussion

- The inheritance of this genetic mutation doesn't guarantee a cancer diagnosis of cancer later in life
- If a risk for inheritance is know genetic testing is extremely important
- There are several screening and prevention options for either those with a positive result or those without
- This mutation isn't as common as some may think

References

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