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Genetic Testing For Cancer

The medical field is a field that is constantly advancing; whether it be with research, testing, or cures, there is always beneficial studies being discovered. Although, genetic testing for different types of cancers is a topic that is developing perhaps more than most. Scientists and doctors are always finding new research to improve the accuracy of tests that can determine if an individual has a gene that dramatically increases their risk for cancer. In order to help prevent cancer, it is imperative to know what genetic testing is, the statistics surrounding the research, and the benefits it can have on one's life.

Cancer is the result of mutations within genes, these mutations can occur one of three ways: sporadic, familial, and hereditary (Adams). The cancer that occurs most often is sporadic cancer, which occurs solely by chance. People who acquire sporadic cancer do not have a family history of that cancer, nor do they have an inherited mutation in their DNA, which would increase their risk for that type of cancer (Definition). Sporadic cancer occurs when certain cells in a person's body develops mutations that later lead to cancer. When a cell acquires mutations that hinder its ability to grow and divide properly, a tumor forms (Our). Familial cancer is the second most common cancer. It is likely caused by the combination of genetic and environmental risk factors. People with familial cancer may have multiple family members with the same type of cancer, although there is not always a specific pattern of inheritance that is traced (Hereditary Colorectal). The reoccurrence of the same cancer within a family can come from shared environmental or lifestyle factors that can be dangerous (Definition). The least common but most predictable type of cancer is hereditary cancer. It is caused by gene mutations that are passed down from parent to child, if a person possesses a certain cancer gene; every cell in their body contains the mutation (Adams). Certain cancer patterns are often seen within families when it comes to hereditary cancer, these patterns can include close family members (for example a mother, daughter, and sister) all being diagnosed with the same type of cancer, family members developing cancer at a young age, or a single family member developing multiple types of cancer (Definiton). Hereditary cancer is the most predictable because of the genetic tests that are available to determine if a person has a genetic mutation that could have been passed down to them from their parents.

Hereditary breast and ovarian cancer is due to an alteration in the *BRAC1* or *BRAC2* gene a vast majority of the time; these gene mutations can be passed down from the mother or the father (Hereditary breast). Inheriting the mutation causes an increased risk not only for breast and ovarian cancer, but also increases the risk for pancreatic and prostate cancer (Testing). There are four types of tests available that look for the *BRCA1* and *BRCA2* genes; the two most common tests are the integrated BRAC*Analysis* and the single site BRAC*Analysis.* The integrated test is for individuals who do not have a family member who has previously been tested, or no one has tested positive for either of the genes. The single site test is for individuals who already know that a gene mutation is present in the family, and the test will examine the individual for that specific mutation (Testing). The two least common tests are the multisite 3 BRAC*Analysis* and the BRAC*Analysis* Large Rearrangement Test. The multisite 3 test examines the three most common mutations found in individuals that come from the Ashkenazi Jewish ancestry. The large rearrangement test, commonly referred to as BART, tests for gene mutations other than *BRCA1* and *BRCA2* that are less common (Testing). To be tested for abnormal gene mutations, an individual should first talk to their doctor about which test is right for them. A blood sample can then be taken by their doctor and sent to a research testing facility or commercial laboratory (Genetic).

After being tested, there are three possible results that a person can receive back: a positive result, a negative result, or an uncertain variant (Inherited breast). A positive result indicates that an individual possesses a *BRCA1* or *BRCA2* mutation and has an increased risk of developing certain types of cancers. Although, just because they have the gene mutation, it is undeterminable if they will ever actually develop cancer (BRCA1). A negative test result is more complex than a positive result and can be more difficult to understand. A negative result always means that the individual tested does not have the *BRCA1* or *BRCA2* gene mutation, but their risk level depends on their family history of cancer. (BRCA1). If an individual has a blood relative who carries the mutation, but they tested negative, then they are at no increased risk for cancer over the general population. Although, the current genetic tests being practiced are not perfect, and the chances of a test missing a harmful mutation is extremely low, but it is possible. For a person who has no identified mutations within their family, but has a family history that suggests the possibility of them carrying the mutation, their negative result dramatically decreases their cancer risk, but not to as low of a risk as the general population (BRCA1). An uncertain variant indicates that an unusual change has been identified in the genes; approximately 2%-4% of people tested receive the uncertain variant result back. It is currently unknown whether these changes are associated with an increased risk for cancer (What to). Although, with new research constantly being

conducted, scientists are learning more about these rare changes, and will soon have a better understanding of what they indicate and how they affect a person's risk for cancer (BRCA1).

About five to ten percent of breast cancers diagnosed are hereditary; therefore the individuals diagnosed are carriers of the BRCA mutations and have significantly increased risks. In general, these risks include being diagnosed at a young age, developing cancer in both breasts, or being diagnosed with multiple types of cancer in a lifetime (Inherited). Individuals with a mutation have up to an 87% chance of developing breast cancer, this is by the age of 70 for women and by age 80 for men. In comparison, the general population only has an 8% chance (Hereditary breast). Along with breast cancer, about five to ten percent of ovarian cancers are also hereditary (Inherited). Carriers of a mutation have up to a 44% chance of being diagnosed with ovarian cancer while the general population has less than a 1% chance (Hereditary breast). Secondary primary breast cancer is when cancer that originally develops in the breasts spreads to other parts of the body, the secondary cancer is made up of breast cancer cells (What is). The risk of secondary primary breast cancer for people who have a *BRCA1* or *BRCA2* gene mutation is up to 64%, while the general populations is only up to 11% (Inherited breast). "This is not a common occurrence, but it does happen (King)".

Other than the *BRAC1* and *BRAC2* gene mutations, there are also mutations within other genes that can lead to cancer. When an individual possesses a mutation within the *MLH1, MSH2, PMS5,* or *EPCAM* gene*;* they are at risk for developing a condition called Lynch syndrome. Lynch syndrome is also referred to as Hereditary Non-polyposis Colorectal Cancer syndrome, and is the most common cause of hereditary colon cancer (Myriad). An individual with Lynch syndrome has an increased risk particularly for colon and rectum cancer, but their risk also increases for cancers of the stomach, small intestine, liver, gallbladder, brain, skin, and cancer of the ovaries for women (Lynch). A person that carries the Lynch mutation has up to an 82% chance of developing colorectal cancer by the age of 70, as opposed to the general population who has only a 2% chance (Myriad). If an individual has a family history of cancers that could be linked to Lynch syndrome, it is recommended that they get tested for a mutation within their genes. The same way one can be tested for a change in *BRAC* genes, one can be tested for a change in their genes that indicate Lynch syndrome; a test for Lynch syndrome has the same three possible results that a *BRACAnalysis* test can have: positive result, negative result, or uncertain variant (Lynch).

Being genetically tested, whether it be for cancer or any other type of medical reason, can have numerous benefits on ones life. When it comes to the mental and emotional benefits, being tested and finding out what risk level a person is at

for developing cancer can reduce anxiety and any stress they may have. It also informs them on the healthiest way to live; a carrier of a mutation can reduce their risk simply by changing certain aspects of their lifestyle. “If you have a strong family history of cancer and test positive, then you control your decisions” (King). Lastly, being tested can provide important information to family members and help them determine what their risk is (Hereditary Breast). There are also limitations when it comes to genetic testing, the most notable being that testing does not detect all causes of hereditary cancer (Hereditary Breast). If a person receives an uncertain variant back from a laboratory as their test result, they are at a disadvantage because it can not be fully said what exactly their gene mutations indicate and what risk they are at for developing cancer.

If an individual is a carrier of a gene mutation, there are multiple things they can do to manage their risk. “You can choose medical management vs. risk-reducing surgery and it is all when you choose to do it” (King). Increased surveillance is perhaps the simplest and easiest way to regulate the uncertainty, monthly breast exams, yearly mammograms and MRI’s, pelvic exams twice a year, and transvaginal ultrasounds can detect cancer at the earlier stages, therefore making it easier to manage. Chemoprevention such as the drug tamoxifen and birth control pills have been shown to reduce the risk of breast cancer and ovarian cancer in women who have certain gene mutations. Tamoxifen can reduce the risk of breast cancer by 53%, and birth control can reduce the risk of ovarian cancer by 60% (Hereditary Breast). Preventive surgery is the type of management that is the most effective. Removing an organ where cancer is likely to develop decreases a women’s risk the most significantly.

When it comes to the cost of testing, prices can end up being very high. Luckily, insurance coverage for testing is excellent and the majority of patients are covered. The average person with insurance pays less than $100. There are two major laws in place for the privacy of patients tested, GINA and HIPPA. These laws prohibit discrimination regarding insurance premiums, health benefits, and employment eligibility when based on a person’s genetic information. After a person has been tested, whether the result is positive or negative, it is important for them to follow through with the next steps. Talking with a genetic counselor and family can help an individual with the coping process. Continuing with testing along with self-examinations is also crucial for remaining healthy and managing a persons cancer risk (Hereditary Breast).

The advancing field of genetic testing is a field that not enough people are aware of. When a form of cancer is common within a family, family members being tested will provide informative information and multiple benefits for a person’s life. In order to help prevent cancer, it is imperative to know what genetic testing is, the statistics surrounding the research, and the benefits it can have on one’s life.

Work Cited

Adams, Amy. "Hereditary Versus Spontaneous Cancer." *Hereditary Versus Spontaneous*

Cancer. Latest Medical Inc., 26 Mar. 2011. Web. 05 Oct. 2015.

"BRCA1 & BRCA2: Cancer Risk & Genetic Testing." *National Cancer Institute*.

National Institutes of Health, n.d. Web. 05 Oct. 2015.

"Definition of Sporadic, familial, hereditary Cancer - NCI Dictionary of Cancer

Terms." *National Cancer Institute*. National Institutes of Health, n.d. Web. 05 Oct. 2015.

"Genetic Testing Facilities and Cost." *Breastcancer.org*. Breastcancer.org, 21 Aug. 2014.

Web. 05 Oct. 2015.

Hereditary Colorectal Cancer. Salt Lake City: Myriad Genetic Labrotories, 2014. Print.

Hereditary Breast and Ovarian Cancer Syndrome. Salt Lake City: Myriad Genetic

Labrotories, 2014. Print.

"Inherited Risk for Breast & Ovarian Cancers." *Hereditary Cancer & Genetics:*.

Memorial Sloan Kettering Cancer Center, 2015. Web. 07 Oct. 2015.

King, Heather. Personal Interview. 2 October 2015.

"Lynch Syndrome." *Genetics Home Reference*. U.S National Library of Medicine, 5 Oct.

2015. Web. 12 Oct. 2015.

"Myriad Genetics | Patients & Families | Hereditary Colon Cancer." *Myriad.com*. Myriad

Genetics, 2015. Web. 12 Oct. 2015.

"Our Doctors." *Sporadic vs. Hereditary Cancer*. Staten Island University

Hospital, n.d. Web. 05 Oct. 2015

"Testing Options." *MySupport360*. Myriad Genetics, 2015. Web. 05 Oct. 2015.

"What Is Secondary Breast Cancer?" *What Is Secondary Breast Cancer?* Cancer

Research UK, 7 Aug. 2014. Web. 07 Oct. 2015.

"What to Do If Your Genetic Test Results Are Uncertain." *Breastcancer.org*. Breastcancer.org, 2015. Web. 07 Oct. 2015.