Genetic counseling and risk assessment for cancer survivors

Melody White, MS, CGC
Genetic Counselor, Cancer Risk Clinic
The University of Chicago Medical Center
773-834-1760

The risk of cancer recurrence is a concern for most cancer survivors. When cancer is possibly caused by an inherited susceptibility other concerns arise. The risk of another new malignancy, the chance of passing on a genetic predisposition to children, the risk for other relatives to develop the same or other cancers, the likelihood of developing treatment-induced infertility and reproductive options are some of the issues that might be discussed during cancer risk assessment and genetic counseling. Patients who have cancer already may incorrectly conclude that learning new information about their genetic status won't change their management since they already developed cancer. In fact, identifying an inherited susceptibility may be life saving for the cancer survivor due to consideration of additional surgery, surveillance, or prevention options. Although healthy individuals who have not had cancer are often referred for genetic counseling and risk assessment, the relative who had cancer is usually the most appropriate person to consider genetic testing when family history is suggestive of an inherited cancer syndrome.

What is genetic counseling and who should consider cancer risk assessment?

Genetic counseling is the provision of information by trained medical professionals who specialize in translating complicated medical and genetic information into understandable terms and concepts to the general population. In the cancer risk setting, this is often provided as part of a team. Patients may meet with an oncologist, geneticist, genetic counselor, nurse, psychologist, and social worker.

Table 1. Genetic counseling and risk assessment is appropriate for individuals who have:

- Early onset cancer
- Multiple cancers
- A strong family history of cancer (similar or various).
- Concerns about treatment induced infertility and assisted reproductive technology options
- Concerns about risk to children (future and existing), or
- Unaffected individuals who are worried about developing cancer for any reason

Additionally, for some women and men with recently diagnosed cancer, genetic counseling and risk assessment may be helpful prior to a final decision regarding treatment and surgery. For example, some women at increased risk for a second breast cancer may choose to have bilateral mastectomy at the time of their original surgery.

Hereditary predisposition to cancer vs. random cancer: how to tell the difference

Only 5%-10% of all cancer is due to an inherited susceptibility. Random cancers generally occur in older individuals without a strong family history of cancer. There are several features of inherited cancers that help the cancer risk team identify high-risk individuals.

Table 2. Features of Hereditary Cancer Syndromes:

- Cancer under the age of 50
- Two or more first-degree relatives with the same type of cancer (i.e. mother and sister)
- Individuals with multiple primary cancers
- Individuals with rare cancers
- Bilateral cancer (i.e. cancer in both breasts)
- Breast and ovarian cancer in a family
- Ashkenazi Jewish ancestry in families with breast and ovarian cancer
- Male breast cancer
- Individuals with multiple adenomatous colon polyps

When is genetic testing offered and what are the risks and benefits?

After genetic counseling and cancer risk assessment, the likelihood of an inherited susceptibility to cancer is determined. The risks, benefits, and limitations of genetic testing are discussed when there are genes that are known to contribute to the types of cancer seen in a family. Whenever possible, it is best to begin by offering genetic testing to a family member who has cancer and is most likely to test positive if a genetic change (mutation) is present. Sometimes when there is no relative with cancer available to test, it is necessary to begin the testing process with an unaffected person. Unfortunately, a negative test result will not be considered a "true" negative unless a mutation is identified in another relative. In such cases, cancer surveillance and surgical recommendations will be based on the patient's personal medical history and family history. The knowledge that a mutation is present in a family can aid in detection of cancer by leading family members to participate in earlier and more thorough screening or can reduce the risk of cancer by leading family members to preventative (prophylactic) surgeries or risk reducing medication. At the same time, relatives of mutation positive individuals can avoid unnecessary cancer screening and prophylactic surgeries. Of course finding out that you are a "true" negative can greatly relieve anxiety about being at increased risk for cancer. Everyone is at risk for cancer so testing negative for a known mutation means they have the same cancer risk as the general population.

Risk to children and when do they need to know about their genetic status?

Most cancer susceptibility syndromes follow an autosomal dominant pattern on inheritance. This means that a single mutation in a gene is inherited causing an increased susceptibility to cancer. Children, siblings and parents of someone with an identified mutation have a 50% chance to have inherited the same mutation. Children should be made aware of their genetic risk when they are emotionally and intellectually mature enough to understand and handle this information. Genetic testing is appropriate in adolescents and younger children only when their medical management would be changed immediately due to risk of cancer during childhood. Genetic counseling for young patients with their parents present is recommended so that they are informed about the outcome of testing and how it will impact their health care.

If you are concerned about your personal or family history of cancer, you can find a genetic counselor in your area by contacting www.nsgc.org or by calling (601) 872-7608.