4 Steps to Making Appropriate Referrals for Cancer Genetic Risk Assessment and Counseling

About 5-10% of all cancers are hereditary, due to germline mutations in any of a number of cancer predisposition genes. An individual with an inherited predisposition is at increased risk for developing specific types of cancer. Cancer tends to develop at an earlier age of onset than is typical (usually defined at or under age 50) and there is an increased risk of developing multiple primary cancers.

Most hereditary cancer syndromes are transmitted in an autosomal dominant fashion with incomplete penetrance. Therefore, a child of an individual with a mutation in a cancer predisposition gene has a 50/50 (1 in 2) chance of inheriting the parent's mutation. Specific management guidelines are available for a number of syndromes. These guidelines aim to decrease the morbidity and mortality associated with hereditary cancer syndromes.

The following guide is intended to help healthcare providers identify their patients at increased risk of hereditary cancer and make appropriate referrals for cancer genetic counseling services. The goal is that appropriate risk identification and referral will help connect patients at risk of hereditary cancer syndromes with the appropriate screening and risk reductions strategies.

Step 1: Collect family medical history information and update the family history annually.

ASK YOUR PATIENT:

- Who in the family has had cancer (relationship to your patient)? Include information about 1st and 2nd degree relatives on both the maternal and paternal side of the family.
  - ◊ 1st degree relatives are siblings, parents, and children
  - ◊ 2nd degree relatives are aunts, uncles, grandparents, half siblings, nieces, and nephews
- What type of cancer or cancers did that person have? (Prompt patient to distinguish between primary cancers versus metastatic disease).
- At around what age was the person diagnosed with cancer?
- Any history of benign tumors or other medical problems?
- Has anyone in the family had genetic testing that revealed a mutation in a cancer predisposition gene? (For this question, you can ask beyond 1st and 2nd degree relatives.) If so, who (relationship to patient) and what gene if known? It can also be helpful to know what laboratory did the testing.

It is important to ask about all cancers in the family- do not limit yourself to a single type of a cancer. Many cancer syndromes are associated with an increased risk of multiple types of cancer.

Asking about the AGE OF ONSET is critical to accurately assessing risk. Any early age of onset (generally ≤ age 50) is a risk factor for hereditary cancer. So even if your patient does not know the exact age of onset, knowing whether the relative was younger or older than 50 can be helpful.

Step 2: Identify personal medical history and family history factors associated with hereditary cancer risk.

GENERAL RISK FACTORS INCLUDE:

- Early age of onset of cancer in your patient (≤50 years of age) or in a first or second degree relative
- Two or more primary tumors or bilateral cancer in paired organs in your patient or in a first or second degree relative (aunts, uncles, grandparents, half siblings, nieces and nephews)
- Rare cancer (e.g., male breast cancer, ovarian cancer, medullary thyroid cancer) in your patient or in a first or second degree relative.
- The same cancer or related cancers (e.g., breast and ovarian cancer, colon and endometrial cancer, colon and ovarian cancer) in 2 or more generations on the same side of the family.
- A 1st or 2nd degree relative with a mutation in a cancer predisposition gene.

Step 3: Talk to your patient about the results of your risk assessment.

It is important to communicate with patients about the significance of their family history whether negative or positive. Example scripts are provided below.

Negative family history (no apparent increased risk): *"I collected your family history because for some people, having a family history of cancer increases their chances of developing certain types of cancer. When that is the case we often offer additional cancer screening or sometimes treatments to prevent cancer. At this time, I did not find anything in your family history that shows you are at increased risk (explain if necessary). But if anything changes in your history, for instance if you have a close relative who is diagnosed with cancer at <50 or someone in your family is found to have a change in a gene associated with an increased risk of cancer, please let me know."*

Positive family history (possible increased risk): "*I collected your family history today because for some people, having a family history of cancer increases their risk of developing certain types of cancers. In your family, the fact that (describe relevant family history factors), makes me concerned that your chance of developing this type(s) of cancer may be increased. If that is the case, you could benefit from increased cancer screening and it might be helpful to talk about ways of preventing cancer. I would like to refer you to a cancer genetics/risk evaluation clinic to review your family history in more detail. The genetic counselor will collect additional family history and discuss with you whether your chance of developing certain types of cancers is increased over that of the average person. S/he may also talk to you about genetic testing for inherited susceptibility to cancer risk if appropriate based on your family history. In that case, it is up to you whether you want to have the test. Whether or not you think you would consider genetic testing, I would like you to see the genetic counselor. The information s/he will collect about your family history will be sent back to me and this will help me make sure that I am managing your cancer risk in the best way possible."*

Step 4: Make appropriate referrals for cancer genetic counseling and testing services .

- **Identify cancer genetics services in your area.** A list of providers in Michigan is available at https://migrc.org/Library/MCGA/MCGADirectory.html
  When local providers are not available, genetic counseling services via telephone are an option. Studies have shown that telephone genetic counseling can be just as effective as in-person services for some patients.
- **Recognize that genetic counseling services are often covered.** This is particularly the case for patients that are at increased risk/meet referral criteria. But like any medical service, the type of policy a patient has will affect coverage. Provide a referral and then encourage the patient to contact the genetic counseling office and their insurance provider if they have any questions about coverage.
  - ◊ For instance, BRCA1/2 counseling and testing is a covered benefit under the Affordable Healthcare Act for women who meet defined criteria. http://www.hhs.gov/healthcare/facts-and-features/fact-sheets/preventive-services-covered-under-aca/
- **Reach out to your genetic counseling service provider.** Genetics providers are usually willing and available to answer your questions about appropriate referrals, the referral process, and coverage. They are also often available to provide your office with educational materials or in person education programs to help you identify your high risk patients.

Primary care providers like you are the gatekeepers for helping patients gain access to appropriate genetic counseling and testing services. **You provide a valuable, potentially life-saving service when you identify patients at increased risk for cancer.** You also provide a valuable service when you can provide reassurance to those patients at population risk. By making the commitment to regularly collect and update family history information, assess and communicate risk to your patients, and make appropriate referrals for cancer genetic counseling, you are giving your patients the best chance to reduce adverse outcomes.