Genetic testing for someone who may be at risk for an inherited disease is always easier if we know the specific genetic cause. Often times, the best way to find the genetic cause is to start by testing someone in the family who is known or strongly suspected to have the disease. If their testing is positive, then we can say that we have found the familial pathogenic (harmful) variant. We can use this as a marker to test other members of the family to see who is also at risk.

An example of this may be a woman who comes in to see a genetic counselor because she has a family history of breast cancer. Her mother had breast cancer at 42, which is considered early-onset, and had recently gotten genetic testing and found that she has a pathogenic variant in the BRCA1 gene, which causes hereditary breast and ovarian cancer syndrome (HBOC). Individuals with HBOC have a higher chance to develop certain types of cancer, particularly breast and ovarian cancers in women. This means that our patient has a 50% chance to also carry that same pathogenic gene variant. Because we were able to find what the specific cause is for the increased cancer risk in her family:

- if our patient tests negative for the familial pathogenic variant, we will be able to tell her that she should have the same risk as the average woman for breast cancer and ovarian, regardless of her family history
- if she tests positive for that pathogenic gene variant, then we know she is at an increased risk for breast and ovarian cancer, and we can discuss different pathways to help keep her healthy

Who the best person to start genetic testing within the family to try to identify a familial pathogenic variant is complex, and depends on what the condition is that you are testing for and the inheritance pattern, as well as your family history. Genetic testing also may not be indicated or helpful, depending on the medical concern.

Testing for a familial pathogenic variant can also be done prenatally, or during pregnancy. If one or both parents are found to be carriers for a genetic condition, a procedure such as a CVS or amniocentesis can be used to get fetal cells that can then be tested. These procedures come with some risk of miscarriage, so it is important to talk with your doctor or genetic counselor to make sure you have all of the information you need to make the decision that is best for you and your family.

A healthcare professional with a background in genetics, such as a genetic counselor, can help you assess all of this information and provide support.

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