

GERMLINE MUTATIONS

Illustrating the Cycle of Hereditary Cancer in Families

Found in every cell in your body, your genes control how your body looks, grows, and functions. You have two copies of every gene. You inherited one copy of every gene you have from your mother and another copy from your father. Among the thousands of genes that you inherited are genes that help prevent tumors and cancers from forming. These are called *tumor suppressor genes*.

Germline mutations are genetic changes that are present in genes in sperm or eggs (germ cells) at the time of conception. Some germline mutations can increase the risk for developing hereditary cancer. Genetic testing looks for changes in genes that have been linked to hereditary cancer. These particular inherited genetic changes are called **pathogenic germline mutations**.

It is estimated that **1 in 279** people carry pathogenic germline mutations that may increase their risk of cancer.

Knowledge is power. Learning if you have an increased risk of cancer because of an inherited genetic mutation can help you take steps to detect cancer at an earlier, more treatable stage or prevent some cancers from ever developing.

This person inherited a germline mutation from a parent.

*As an example, one of the two **BRCA2** tumor suppressor genes this person inherited has a pathogenic mutation.*



*However, the other inherited **BRCA2** gene **does not** have a mutation.*

*That means **every** child of this person has a **50/50** chance of inheriting either the mutated gene or the normal gene.*



*Both **BRCA2** genes this child inherited are normal. There is no mutated **BRCA2** gene to be passed on to future generations. The cycle ends here.*

This child inherited the germline mutation.

*Every child of this person has a **50%** chance of inheriting the germline mutation.*

*Again, **every** child of this person has a **50%** chance of inheriting the germline mutation.*

This person inherited the mutation.

*This individual did not inherit the mutated **BRCA2** gene. **Germline mutations do not skip generations.** The cycle ends here.*

This information has been reviewed by a Certified Genetic Counselor.

This individual did not inherit the mutation. The cycle ends here.

This person inherited the mutation.

*** Breaking the cycle begins with **cascade testing**.** It is the *testing-sharing-testing-sharing* of genetic information in families with a history of cancer. It can identify who is and who isn't at risk for hereditary cancer among family members and help prevent cancer and save lives.

The cycle continues. *

For more information about genetic mutations and hereditary cancer, go to **www.genetionary.org**