

About *CDH1* Gene Mutations

About Genes

Genes are in every cell in our bodies. Genes are made of DNA, which gives instructions to cells about how to grow and work together. We have two copies of each gene in each cell—one from our mother and one from our father. When genes work properly, they help keep cancer cells from developing. If one copy of a gene has a mutation, the gene cannot work correctly. This raises the risk for certain cancers.

The *CDH1* gene has two main jobs. The first is to help cells stick to one another to form tissues. The second is to help prevent cancer from forming.

If you have a mutation in one copy of the *CDH1* gene, it cannot perform its jobs as it normally would. Individuals who inherit mutations in *CDH1* may have inherited one of these hereditary cancer syndromes: *Hereditary Diffuse Gastric Cancer* or *Hereditary Lobular Breast Cancer*.

Hereditary Diffuse Gastric Cancer (HDGC)

HDGC is passed down through families and increases the risk for getting certain types of stomach and breast cancer. The average age people with HDGC get cancer is 37 years old.

Hereditary Lobular Breast Cancer (HLBC)

HLBC is also passed down through families and increases the risk for getting a certain type of breast cancer. Individuals with HLBC have no personal or family history of stomach cancer. The average age people with HLBC get breast cancer is 53 years old.

CDH1 Mutations and Cancer Risks

Stomach Cancer

The type of stomach cancer people with HDGC can get is called diffuse gastric cancer. This cancer grows in the lining of the stomach and is hard to detect.

The risk of stomach cancer for men and women by the age of 80 years old is 70-80% (7-8 in 10 people).

Recommendations

Women and Men

Diffuse gastric cancer is hard to detect. Symptoms of diffuse gastric cancer may not appear until the condition is more severe. For people with HDGC we recommend considering a prophylactic total gastrectomy (removal of the stomach) between the ages of 18-40. If this is not something an individual chooses to do, their doctor may recommend screening by stomach endoscopy. This screening includes taking small samples of the stomach which can be tested for cancer. This screening may start 5-10 years before the age of the first stomach cancer diagnosis in the family. These should be done at least once a year. Unfortunately, it is very difficult to detect diffuse gastric cancer with endoscopy.

Women

At 30 years of age women with HDGC or HLBC should start breast imaging. This should be an annual mammogram and an annual breast MRI. Breast MRI may be better at finding lobular breast cancer.

There are also some medicines that can lower the risk of getting breast cancer. Some women will consider surgery to remove both breasts especially if they have a strong family history of breast cancer.

Children and Siblings

Children and siblings of individuals with *CDH1* mutations have a 1 in 2 or 50% chance of also carrying a *CDH1* mutation. Genetic counseling and testing are recommended for these individuals after the age of 18. A genetic counselor can help give you ideas on how to tell other family members about HDGC or HLBC.

It is important to know which side of the family carries the *CDH1* mutation. This allows those relatives to know about their cancer risk. A genetic counselor can help you know who in your family should be tested.

Family Members Who Test Negative

Family members who test negative for a *CDH1* mutation are at the average population risk for stomach and breast cancer. They should follow general cancer screening guidelines.

Breast Cancer

The type of breast cancer women with both HDGC and HLBC can get starts in the milk producing glands and is called lobular breast cancer.

The risk for women with HDGC or HLBC to get lobular breast cancer in their lifetime is around 40-50% (4-5 in 10 women).

Do you have questions about your risk for cancer? Our doctors and genetic counselors can help find the cancer screening plan you need. Call Huntsman Cancer Institute's Family Cancer Assessment Clinic to learn more: 801-587-9555. Huntsmancancer.org/fcac