

Hereditary Breast/Ovarian Cancer Genetic Testing Frequently Asked Questions

How common are *BRCA1* and *BRCA2* mutations in the general population?

Inherited mutations in *BRCA1* and *BRCA2* are relatively uncommon in the general population. The carrier frequency is estimated to be approximately 1 in 800. Certain ethnic groups, such as Ashkenazi Jews have been shown to have a higher carrier frequency (1 in 40).

What percentage of breast and ovarian cancer cases are due to *BRCA1* and *BRCA2* mutations?

As much as 5-10% of all breast cancer cases and 10% of all ovarian cancer cases are thought to be caused by *BRCA1* and *BRCA2* mutations. Most breast cancer is sporadic (~70%) and as much as 25% is familial, caused by a combination of hereditary and environmental factors. Although 90% of hereditary breast cancer is due to mutations in *BRCA1/2*, other genes have been implicated in some families.

Can *BRCA1* and *BRCA2* mutations be inherited from either side of the family?

Yes, a person can inherit a *BRCA1* or *BRCA2* mutation from either their mother or father.

When should a patient be referred for a risk assessment with a cancer genetics professional to discuss the option of genetic testing for *BRCA1* and *BRCA2* mutations?*

Patients with a personal or family history of:

- Breast cancer diagnosed under age 50

- Bilateral or ipsilateral breast cancer diagnosed at any age

- Ovarian cancer diagnosed at any age

- Breast and ovarian cancer in the same individual

- Male breast cancer

- Three or more family members diagnosed with breast and/or ovarian cancer (at any age)

- Breast or ovarian cancer diagnosed at any age in a family of Ashkenazi (Eastern European) Jewish descent

- Known *BRCA1/2* mutation

*For suggestions on triaging individuals for testing in your own office versus referring for genetic counseling, see the attached triage document.

**The criteria above are to be used as a guide and are not a substitute for clinical judgment. Not all clinical scenarios can be anticipated.

What are the benefits of genetic counseling when offering *BRCA1* and *BRCA 2* testing?

Cancer genetics professionals are trained to evaluate family history for hereditary cancer syndromes of all types and offer genetic testing that is appropriate. Patient education and informed consent are critical aspects of the genetic testing process and will help individuals decide if genetic testing is right for them. Pretest counseling performed by a genetics professional addresses the implications of potential test results, medical management, risks to family members, psychosocial aspects, other options for testing and testing costs. If help is needed, a genetics professional can also provide assistance with dissemination of relevant information to at-risk family members.

How much does BRCA1 and BRCA2 genetic testing cost?

Comprehensive analysis of the BRCA1 and BRCA2 genes	\$3,120
Single site testing for a known mutation in the family	\$440
Ashkenazi Jewish multi-site 3 testing	\$535

Other testing options exist at varying costs. While genetic testing is generally covered by insurance, this may be dependent on the insurance company, patient deductibles and coinsurance, meeting medical criteria and other factors.

Will my patient be discriminated based on genetic test results?

Despite concerns in the past about possible problems arising from genetic testing, current state and federal laws make it illegal for health insurers and employers to discriminate against a person based on their genetic status.

What is the risk for breast and ovarian cancer in individuals found to have a BRCA1 or BRCA2 mutation?

For women with a mutation in either *BRCA1* or *BRCA2*, studies have shown the lifetime risk of breast cancer to range between 50-87%. The lifetime risk of ovarian cancer is estimated to range between 27-44%, depending on the specific mutation. Men with a *BRCA* mutation have a 5-10 percent lifetime risk of developing breast cancer and a 20% lifetime risk for prostate cancer.