BRCA1 and *BRCA2*: Cancer Risk and Genetic Testing

Key Points

- *BRCA1* and *BRCA2* are human genes that belong to a class of genes known as tumor suppressors. Mutation of these genes has been linked to hereditary breast and ovarian cancer (see Question 1).
- A woman's risk of developing breast and/or ovarian cancer is greatly increased if she inherits a deleterious (harmful) *BRCA1* or *BRCA2* mutation. Men with these mutations also have an increased risk of breast cancer. Both men and women who have harmful *BRCA1* or *BRCA2* mutations may be at increased risk of other cancers (see Question 2).
- Genetic tests are available to check for *BRCA1* and *BRCA2* mutations. A blood sample is required for these tests, and genetic counseling is recommended before and after the tests (see Question 5).
- If a harmful *BRCA1* or *BRCA2* mutation is found, several options are available to help a person manage their cancer risk (see Question 11).
- Federal and state laws help ensure the privacy of a person's genetic information and provide protection against discrimination in health insurance and employment practices (see Questions 14 and 15).
- Many research studies are being conducted to find newer and better ways of detecting, treating, and preventing cancer in *BRCA1* and *BRCA2* mutation carriers. Additional studies are focused on improving genetic counseling methods and outcomes. Our knowledge in these areas is evolving rapidly (see Question 18).

1. What are *BRCA1* and *BRCA2*?

BRCA1 and *BRCA2* are human genes that belong to a class of genes known as tumor suppressors.

In normal cells, *BRCA1* and *BRCA2* help ensure the stability of the cell's genetic material (DNA) and help prevent uncontrolled cell growth. Mutation of these genes has been linked to the development of hereditary breast and ovarian cancer.

The names *BRCA1* and *BRCA2* stand for **br**east **ca**ncer susceptibility gene **1** and **br**east **ca**ncer susceptibility gene **2**, respectively.

2. How do *BRCA1* and *BRCA2* gene mutations affect a person's risk of cancer?

Not all gene changes, or mutations, are deleterious (harmful). Some mutations may be beneficial, whereas others may have no obvious effect (neutral). Harmful mutations can increase a person's risk of developing a disease, such as cancer.

A woman's lifetime risk of developing breast and/or ovarian cancer is greatly increased if she inherits a harmful mutation in *BRCA1* or *BRCA2*. Such a woman has an increased risk of developing breast and/or ovarian cancer at an early age (before menopause) and often has multiple, close family members who have been diagnosed with these diseases. Harmful *BRCA1* mutations may also increase a woman's risk of developing cervical, uterine, pancreatic, and colon cancer (1, 2). Harmful *BRCA2* mutations may additionally increase the risk of pancreatic cancer, stomach cancer, gallbladder and bile duct cancer, and melanoma (3).

Men with harmful *BRCA1* mutations also have an increased risk of breast cancer and, possibly, of pancreatic cancer, testicular cancer, and early-onset prostate cancer. However, male breast cancer, pancreatic cancer, and prostate cancer appear to be more strongly associated with *BRCA2* gene mutations (2–4).

The likelihood that a breast and/or ovarian cancer is associated with a harmful mutation in *BRCA1* or *BRCA2* is highest in families with a history of multiple cases of breast cancer, cases of both breast and ovarian cancer, one or more family members with two primary cancers (original tumors that develop at different sites in the body), or an Ashkenazi (Central and Eastern European) Jewish background (see Question 6). However, **not every** woman in such families carries a harmful *BRCA1* or *BRCA2* mutation, and **not every** cancer in such families is linked to a harmful mutation in one of these genes. Furthermore, **not every** woman who has a harmful *BRCA1* or *BRCA2* mutation will develop breast and/or ovarian cancer.

According to estimates of lifetime risk, about 12.0 percent of women (120 out of 1,000) in the general population will develop breast cancer sometime during their lives compared with about 60 percent of women (600 out of 1,000) who have inherited a harmful mutation in *BRCA1* or *BRCA2* (4, 5). In other words, a woman who has inherited a harmful mutation in *BRCA1* or *BRCA2* is about five times more likely to develop breast cancer than a woman who does not have such a mutation.

Lifetime risk estimates for ovarian cancer among women in the general population indicate that 1.4 percent (14 out of 1,000) will be diagnosed with ovarian cancer compared with 15 to 40 percent of women (150–400 out of 1,000) who have a harmful *BRCA1* or *BRCA2* mutation (4, 5).

It is important to note, however, that most research related to *BRCA1* and *BRCA2* has been done on large families with many individuals affected by cancer. Estimates of breast and ovarian cancer risk associated with *BRCA1* and *BRCA2* mutations have been calculated from studies of these families. Because family members share a proportion of their genes and, often, their environment, it is possible that the large number of cancer cases seen in these families may be due in part to other genetic or environmental factors. Therefore, risk estimates that are based on families with many affected members may not accurately reflect the levels of risk for *BRCA1* and *BRCA2* mutation carriers in the general population. In addition, no data are available from long-term studies of the general population comparing cancer risk in women who have harmful *BRCA1* or *BRCA2* mutations with women who do not have such mutations. Therefore, the percentages given above are estimates that may change as more data become available.

3. Do inherited mutations in other genes increase the risk of breast and/or ovarian tumors?

Yes. Mutations in several other genes, including *TP53*, *PTEN*, *STK11/LKB1*, *CDH1*, *CHEK2*, *ATM*, *MLH1,* and *MSH2*, have been associated with hereditary breast and/or ovarian tumors (4, 6, 7). However, the majority of hereditary breast cancers can be accounted for by inherited mutations in *BRCA1* and *BRCA*2 (8). Overall, it has been estimated that inherited *BRCA1* and *BRCA2* mutations account for 5 to 10 percent of breast cancers and 10 to 15 percent of ovarian cancers among white women in the United States (6).

4. Are specific mutations in *BRCA1* and *BRCA2* more common in certain populations?

Yes. For example, three specific mutations, two in the *BRCA1* gene and one in the *BRCA2* gene, are the most common mutations found in these genes in the Ashkenazi Jewish population. In one study, 2.3 percent of participants (120 out of 5,318) carried one of these three mutations (9). This frequency is about five times higher than that found in the general population (10). It is not known whether the increased frequency of these mutations is responsible for the increased risk of breast cancer in Jewish populations compared with non-Jewish populations.

Other ethnic and geographic populations around the world, such as the Norwegian, Dutch, and Icelandic peoples, also have higher frequencies of specific *BRCA1* and *BRCA2* mutations.

In addition, limited data indicate that the frequencies of specific *BRCA1* and *BRCA2* mutations may vary among individual racial and ethnic groups in the United States, including African Americans, Hispanics, Asian Americans, and non-Hispanic whites (11–13).

This information about genetic differences between racial and ethnic groups may help health care providers in selecting the most appropriate genetic test(s) (see Question 5).

5. Are genetic tests available to detect *BRCA1* and *BRCA2* mutations, and how are they performed?

Yes. Several methods are available to test for *BRCA1* and *BRCA2* mutations (14). Most of these methods look for changes in *BRCA1* and *BRCA2* DNA. At least one method looks for changes in the proteins produced by these genes. Frequently, a combination of methods is used.

A blood sample is needed for these tests. The blood is drawn in a laboratory, doctor's office, hospital, or clinic and then sent to a laboratory that specializes in the tests. It usually takes several weeks or longer to get the test results. Individuals who decide to get tested should check with their health care provider to find out when their test results might be available.

Genetic counseling is generally recommended before and after a genetic test. This counseling should be performed by a health care professional who is experienced in cancer genetics (see Question 17). Genetic counseling usually involves a risk assessment based on the individual's personal and family medical history and discussions about the appropriateness of genetic testing, the specific test(s) that might be used and the technical accuracy of the test(s), the medical implications of a positive or a negative test result, the possibility that a test result might not be informative (an ambiguous result) (see below), the psychological risks and benefits of genetic test results, and the risk of passing a mutation to children.

6. How do people know if they should consider genetic testing for *BRCA1* and *BRCA2* mutations?

Currently, there are no standard criteria for recommending or referring someone for *BRCA1* or *BRCA2* mutation testing.

In a family with a history of breast and/or ovarian cancer, it may be most informative to first test a family member who has breast or ovarian cancer. If that person is found to have a harmful *BRCA1* or *BRCA2* mutation, then other family members can be tested to see if they also have the mutation.

Regardless, women who have a relative with a harmful *BRCA1* or *BRCA2* mutation and women who appear to be at increased risk of breast and/or ovarian cancer because of their family history should consider genetic counseling to learn more about their potential risks and about *BRCA1* and *BRCA2* genetic tests.

The likelihood of a harmful mutation in *BRCA1* or *BRCA2* is increased with certain familial patterns of cancer. These patterns include the following (15):

- For women who are not of Ashkenazi Jewish descent:
  - two first-degree relatives (mother, daughter, or sister) diagnosed with breast cancer, one of whom was diagnosed at age 50 or younger;
  - three or more first-degree or second-degree (grandmother or aunt) relatives diagnosed with breast cancer regardless of their age at diagnosis;
  - a combination of first- and second-degree relatives diagnosed with breast cancer and ovarian cancer (one cancer type per person);
  - a first-degree relative with cancer diagnosed in both breasts (bilateral breast cancer);
  - a combination of two or more first- or second-degree relatives diagnosed with ovarian cancer regardless of age at diagnosis;
  - a first- or second-degree relative diagnosed with both breast and ovarian cancer regardless of age at diagnosis; and
  - breast cancer diagnosed in a male relative.

- For women of Ashkenazi Jewish descent:
  - any first-degree relative diagnosed with breast or ovarian cancer; and
  - two second-degree relatives on the same side of the family diagnosed with breast or ovarian cancer.

These family history patterns apply to about 2 percent of adult women in the general population. Women who have none of these family history patterns have a low probability of having a harmful *BRCA1* or *BRCA2* mutation.

7. How much does *BRCA1* and *BRCA2* mutation testing cost?

The cost for *BRCA1* and *BRCA2* mutation testing usually ranges from several hundred to several thousand dollars. Insurance policies vary with regard to whether or not the cost of testing is covered. People who are

considering *BRCA1* and *BRCA2* mutation testing may want to find out about their insurance company's policies regarding genetic tests.

8. What does a positive *BRCA1* or *BRCA2* test result mean?

A positive test result generally indicates that a person has inherited a known harmful mutation in *BRCA1* or *BRCA2* and, therefore, has an increased risk of developing certain cancers, as described above. However, a positive test result provides information only about a person's **risk** of developing cancer. It cannot tell whether an individual will actually develop cancer or when. **Not all** women who inherit a harmful *BRCA1* or *BRCA2* mutation will develop breast or ovarian cancer.

A positive genetic test result may have important health and social implications for family members, including future generations. Unlike most other medical tests, genetic tests can reveal information not only about the person being tested but also about that person's relatives. Both men and women who inherit harmful *BRCA1* or *BRCA2* mutations, whether they develop cancer themselves or not, may pass the mutations on to their sons and daughters. However, **not all** children of people who have a harmful mutation will inherit the mutation.

9. What does a negative *BRCA1* or *BRCA2* test result mean?

How a negative test result will be interpreted depends on whether or not someone in the tested person's family is known to carry a harmful *BRCA1* or *BRCA2* mutation. If someone in the family has a known mutation, testing other family members for the same mutation can provide information about their cancer risk. If a person tests negative for a known mutation in his or her family, it is unlikely that they have an inherited susceptibility to cancer associated with *BRCA1* or *BRCA2*. Such a test result is called a "true negative." Having a true negative test result does not mean that a person will not develop cancer; it means that the person's risk of cancer is probably the same as that of people in the general population.

In cases in which a family has a history of breast and/or ovarian cancer and no known mutation in *BRCA1* or *BRCA2* has been previously identified, a negative test result is not informative. It is not possible to tell whether an individual has a harmful *BRCA1* or *BRCA2* mutation that was not detected by testing (a "false negative") or whether the result is a true negative. In addition, it is possible for people to have a mutation in a gene other than *BRCA1* or *BRCA2* that increases their cancer risk but is not detectable by the test(s) used.

10. What does an ambiguous *BRCA1* or *BRCA2* test result mean?

If genetic testing shows a change in *BRCA1* or *BRCA2* that has not been previously associated with cancer in other people, the person's test result may be interpreted as "ambiguous" (uncertain). One study found that 10 percent of women who underwent *BRCA1* and *BRCA2* mutation testing had this type of ambiguous result (16).

Because everyone has genetic differences that are not associated with an increased risk of disease, it is sometimes not known whether a specific DNA change affects a person's risk of developing cancer. As more research is conducted and more people are tested for *BRCA1* or *BRCA2* changes, scientists will learn more about these changes and cancer risk.

11. What are the options for a person who has a positive test result?

Several options are available for managing cancer risk in individuals who have a harmful *BRCA1* or *BRCA2* mutation. However, high-quality data on the effectiveness of these options are limited.

- **Surveillance—**Surveillance means cancer screening, or a way of detecting the disease early. Screening does not, however, change the risk of developing cancer. The goal is to find cancer early, when it may be most treatable.

  Surveillance methods for breast cancer may include mammography and clinical breast exams. Studies are currently under way to test the effectiveness of other breast cancer screening methods, such as magnetic resonance imaging (MRI), in women with *BRCA1* or *BRCA2* mutations. With careful surveillance, many breast cancers will be diagnosed early enough to be successfully treated.

For ovarian cancer, surveillance methods may include transvaginal ultrasound, blood tests for CA–125 antigen, and clinical exams. Surveillance can sometimes find ovarian cancer at an early stage, but it is uncertain whether these methods can help reduce a woman's chance of dying from this disease.

- **Prophylactic Surgery—**This type of surgery involves removing as much of the "at-risk" tissue as possible in order to reduce the chance of developing cancer. Bilateral prophylactic mastectomy (removal of healthy breasts) and prophylactic salpingo-oophorectomy (removal of healthy fallopian tubes and ovaries) do not, however, offer a guarantee against developing cancer. Because not all at-risk tissue can be removed by these procedures, some women have developed breast cancer, ovarian cancer, or primary peritoneal carcinomatosis (a type of cancer similar to ovarian cancer) even after prophylactic surgery. In addition, some evidence suggests that the amount of protection salpingo-oophorectomy provides against the development of breast and ovarian cancer may differ between carriers of *BRCA1* and *BRCA2* mutations (17).

- **Risk Avoidance—**Certain behaviors have been associated with breast and ovarian cancer risk in the general population (see Question 16). Research results on the benefits of modifying individual behaviors to reduce the risk of developing cancer among *BRCA1* or *BRCA2* mutation carriers are limited.

- **Chemoprevention—**This approach involves the use of natural or synthetic substances to reduce the risk of developing cancer or to reduce the chance that cancer will come back. For example, the drug tamoxifen has been shown in numerous clinical studies to reduce the risk of developing breast cancer by about 50 percent in women who are at increased risk of this disease and to reduce the recurrence of breast cancer in women undergoing treatment for a previously diagnosed breast tumor. As a result, tamoxifen was approved by the U.S. Food and Drug Administration (FDA) as a breast cancer treatment and to reduce the risk of breast cancer development in premenopausal and postmenopausal women who are at increased risk of this disease. Few studies, however, have evaluated the effectiveness of tamoxifen in women with *BRCA1* or *BRCA2* mutations. Data from three studies suggest that tamoxifen may be able to help lower the risk of breast cancer in *BRCA1* and *BRCA2* mutation carriers (18–20). Two of these studies examined the effectiveness of tamoxifen in helping to reduce the development of cancer in the opposite breast of women undergoing treatment for an initial breast cancer (19, 20).

  Another drug, raloxifene, was shown in a large clinical trial sponsored by the National Cancer Institute (NCI) to reduce the risk of developing invasive breast cancer in postmenopausal women at increased risk of this disease by about the same amount as tamoxifen. As a result, raloxifene was approved by the FDA for breast cancer risk reduction in postmenopausal women. Since tamoxifen and raloxifene inhibit the growth of breast cancer cells in similar ways, raloxifene may be able to help reduce breast cancer risk in postmenopausal *BRCA1* and *BRCA2* mutation carriers. However, this has not been studied directly.

12. What are some of the benefits of genetic testing for breast and ovarian cancer risk?

There can be benefits to genetic testing, whether a person receives a positive or a negative result. The potential benefits of a negative result include a sense of relief and the possibility that special preventive checkups, tests, or surgeries may not be needed. A positive test result can bring relief from uncertainty and allow people to make informed decisions about their future, including taking steps to reduce their cancer risk. In addition, many people who have a positive test result may be able to participate in medical research that could, in the long run, help reduce deaths from breast cancer.

13. What are some of the risks of genetic testing for breast and ovarian cancer risk?

The direct medical risks, or harms, of genetic testing are very small, but test results may have an effect on a person's emotions, social relationships, finances, and medical choices.

People who receive a positive test result may feel anxious, depressed, or angry. They may choose to undergo preventive measures, such as prophylactic surgery, that have serious long-term implications and whose effectiveness is uncertain.

People who receive a negative test result may experience "survivor guilt," caused by the knowledge that they likely do not have an increased risk of developing a disease that affects one or more loved ones.

Because genetic testing can reveal information about more than one family member, the emotions caused by test results can create tension within families. Test results can also affect personal choices, such as marriage

and childbearing. Issues surrounding the privacy and confidentiality of genetic test results are additional potential risks (see below).

14. What can happen when genetic test results are placed in medical records?

Clinical test results are normally included in a person's medical records. Consequently, individuals considering genetic testing must understand that their results might not be kept private.

Because a person's genetic information is considered health information, it is covered by the Privacy Rule of the Health Information Portability and Accountability Act (HIPAA) of 1996 (21). The Privacy Rule requires that health care providers and others protect the privacy of health information, sets boundaries on the use and release of health records, and empowers individuals to control certain uses and disclosures of their health-related information. Many states also have laws to protect the privacy and limit the release of genetic and other health information.

In 2008, the Genetic Information Nondiscrimination Act (GINA) became Federal law (see Question 15). GINA prohibits discrimination based on genetic information in relation to health insurance and employment, but the law does not cover life insurance, disability insurance, and long-term care insurance. When applying for these types of insurance, people may be asked to sign forms that give an insurance company permission to access their medical records. The insurance company may take genetic test results into account when making decisions about coverage.

Some physicians keep genetic test results out of medical records. However, even if such results are not included in a person's medical records, information about a person's genetic profile can sometimes be gathered from that person's family medical history.

15. What is genetic discrimination, and are there laws to protect people from this type of discrimination?

Genetic discrimination occurs when people are treated differently by insurance companies or employers because they have a gene mutation that increases their risk of a disease, such as cancer. However, in 2008, GINA was enacted to protect U.S. citizens against discrimination based on their genetic information in relation to health insurance and employment (22, 23). The parts of the law relating to health insurers will take effect between May 2009 and May 2010, and those relating to employers will take effect by November 2009. The law does not cover life insurance, disability insurance, and long-term care insurance. In addition, the law does not cover members of the military.

Some of the protections under GINA with regard to health insurance include the following:

- Premiums or contributions to a group health plan cannot be increased based on the genetic information of an individual(s) enrolled in the plan.
- Insurers cannot require an individual or family member to undergo a genetic test before enrollment in a group health plan.
- Insurers cannot request, require, or purchase genetic information about an individual before the person's enrollment in a group health plan or in connection with that person's enrollment in the plan.
- Health insurers cannot use genetic information as the only basis upon which to claim a pre-existing condition is present and, therefore, to deny coverage.

Some of the protections under GINA with regard to employment include the following:

- Employers cannot refuse to hire and cannot fire individuals based on their genetic information.
- Employers cannot discriminate against employees with regard to salary, terms and conditions of employment, privileges, and opportunities for the future because of their genetic information.
- Employers cannot request, require, or purchase genetic information about an employee except under specific circumstances.

- Employers cannot disclose an employee's genetic information except under specific circumstances.

Before GINA was passed, many states enacted laws against genetic discrimination. The amount of protection provided by these laws varies widely from state to state. GINA sets a minimum standard of protection that must be met by all states. It does not weaken the protections provided by any state law.

16. In general, what factors increase or decrease the chance of developing breast cancer and/or ovarian cancer?

The following factors have been associated with increased or decreased risk of developing breast and/or ovarian cancer in the general population. It is not yet known exactly how these factors influence risk in people with *BRCA1* or *BRCA2* mutations. In addition, a significant portion of hereditary breast cancers are not associated with *BRCA1* or *BRCA2* mutations (8).

- **Age—**The risks of breast and ovarian cancer increase with age. Most breast and ovarian cancers occur in women over the age of 50. Women with harmful *BRCA1* or *BRCA2* mutations often develop breast or ovarian cancer **before** age 50.

- **Family History—**Women who have a first-degree relative (mother, sister, or daughter) or other close relative with breast and/or ovarian cancer may be at increased risk of developing these cancers. In addition, women with relatives who have had colon cancer may be at increased risk of developing ovarian cancer.

- **Medical History—**Women who have already had breast cancer are at increased risk of developing breast cancer again, or of developing ovarian cancer.

- **Hormonal Influences—**Estrogen is a hormone that is naturally produced by the body and stimulates the normal growth of breast tissue. It is thought that excess estrogen may contribute to breast cancer risk because of its natural role in stimulating breast cell growth. Women who had their first menstrual period before the age of 12 or experienced menopause after age 55 have a slightly increased risk of breast cancer, as do women who had their first child after age 30. Each of these factors increases the amount of time a woman's body is exposed to estrogen. Removal of a woman's ovaries, which are the main source of estrogen production, reduces the risk of breast cancer. Breast-feeding also reduces breast cancer risk and is thought to exert its effects through hormonal mechanisms (24).

- **Birth Control Pills (Oral Contraceptives)—**Most studies have shown a slight increase or no change in risk of breast cancer among women taking birth control pills (24). In contrast, numerous studies have shown that taking birth control pills decreases a woman's risk of developing ovarian cancer (25). This protective benefit appears to increase with the duration of oral contraceptive use and persists up to 25 years after discontinuing use. It also appears that the use of birth control pills lowers the risk of ovarian cancer in women who carry harmful *BRCA1* or *BRCA2* mutations (26).

- **Hormone Replacement Therapy—**Doctors may prescribe hormone replacement therapy (HRT) to reduce the discomfort of certain symptoms of menopause, such as hot flashes. However, the results of the Women's Health Initiative (WHI), a large clinical study conducted by the National Heart, Lung, and Blood Institute, part of the National Institutes of Health (NIH), showed that HRT with the hormones estrogen and progestin is associated with harmful side effects, including an increased risk of breast cancer and increased risks of heart attack, blood clots, and stroke. The WHI also showed that HRT with estrogen alone was associated with increased risks of blood clots and stroke, but the effect on breast cancer risk was uncertain (27). In addition, the WHI showed an increase in ovarian cancer risk among women who received estrogen and progestin HRT, but this finding was not statistically significant (28). Because of these potential harmful side effects, the FDA has recommended that HRT be used only at the lowest doses for the shortest period of time needed to achieve treatment goals.

  No data have been reported to date regarding the effects of HRT on breast cancer risk among women carrying harmful *BRCA1* or *BRCA2* mutations, and only limited data are available regarding HRT use and ovarian cancer risk among such women. In one study, HRT use did not appear to affect ovarian cancer risk among women with *BRCA1* or *BRCA2* mutations (29).

  When considering HRT use, both the potential harms and benefits of this type of treatment should be discussed carefully by a woman and her health care provider.

- **Obesity—**Substantial evidence indicates that obesity is associated with an increased risk of breast cancer, especially among postmenopausal women who have not used HRT (24). Evidence also suggests that obesity is associated with increased mortality (death) from ovarian cancer (30).

- **Physical Activity—**Numerous studies have examined the relationship between physical activity and breast cancer risk, and most of these studies have shown that physical activity, especially strenuous physical activity, is associated with reduced risk. This decrease in risk appears to be more pronounced in premenopausal women and women with lower-than-normal body weight (24).

- **Alcohol—**There is substantial evidence that alcohol consumption is associated with increased breast cancer risk. However, it is uncertain whether reducing alcohol consumption would decrease breast cancer risk (24).

- **Dietary Fat—**Although early studies suggested a possible association between a high-fat diet and increased breast cancer risk, more recent studies have been inconclusive. In the WHI, a low-fat diet did not help reduce breast cancer risk (31).

17. Where can people get more information about genetic testing for cancer risk?

A person who is considering genetic testing should speak with a professional trained in genetics before deciding whether to be tested. These professionals may include doctors, genetic counselors, and other health care workers trained in genetics (such as nurses, psychologists, or social workers). For help finding a health care professional trained in genetics, please visit NCI's Cancer Genetics Services Directory at http://www.cancer.gov/search/geneticsservices on the Internet. Alternatively, please contact NCI's Cancer Information Service (CIS) (see below for contact information). The CIS can provide more information about genetic testing and help in finding a health care professional trained in genetics.

18. What research is currently being done to help individuals with harmful *BRCA1* or *BRCA2* mutations?

Research studies are being conducted to find newer and better ways of detecting, treating, and preventing cancer in *BRCA1* and *BRCA2* mutation carriers. Additional studies are focused on improving genetic counseling methods and outcomes. Our knowledge in these areas is evolving rapidly.

Information about active clinical trials (research studies with people) for individuals with *BRCA1* or *BRCA2* mutations is available on NCI's Web site. The following links will initiate searches of NCI's clinical trials database and retrieve lists of trials open to individuals with *BRCA1* or *BRCA2* mutations.

- *BRCA1* mutation carriers
- *BRCA2* mutation carriers

In addition, NCI's CIS can provide information about clinical trials and help with clinical trial searches (see below for contact information).

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Related NCI materials and Web pages:

- Cancer Genetics Home Page (http://www.cancer.gov/cancertopics/prevention-genetics-causes/genetics)
- *What You Need To Know About™ Breast Cancer* (http://www.cancer.gov/cancertopics/wyntk/breast)
- *What You Need To Know About™ Ovarian Cancer* (http://www.cancer.gov/cancertopics/wyntk/ovary)
- *Genetic Testing for Breast and Ovarian Cancer Risk: It's Your Choice* (http://www.cancer.gov/cancertopics/Genetic-Testing-for-Breast-and-Ovarian-Cancer-Risk)

How can we help?

We offer comprehensive research-based information for patients and their families, health professionals, cancer researchers, advocates, and the public.

- ***Call*** NCI's Cancer Information Service at 1–800–4–CANCER (1–800–422–6237)
- ***Visit*** us at http://www.cancer.gov or http://www.cancer.gov/espanol
- ***Chat*** using LiveHelp, NCI's instant messaging service, at http://www.cancer.gov/livehelp
- ***E-mail*** us at cancergovstaff@mail.nih.gov
- ***Order*** publications at http://www.cancer.gov/publications or by calling 1–800–4–CANCER
- ***Get help*** with quitting smoking at 1–877–44U–QUIT (1–877–448–7848)

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