

# Women's Health

# Update

Winter 2010

Bureau of Health Promotion and Risk Reduction, Office of Healthy Ohio, Ohio Department of Health



Update Focus: Women and Chronic Diseases



## Genetics and Women's Health

Information on this page can be found in the print version of *Women's Health USA 2008*. U.S. Department of Health and Human Services, Health Resources and Services Administration, Maternal and Child Health Bureau. *Women's Health USA 2008*. Rockville, Maryland: U.S. Department of Health and Human Services, 2008.

### Letter from Debra Seltzer

The topic of this issue, women and genetic diseases, is one that I know little about and suspect most other people don't either. We do hear more and more about the capability of genetics to explain and predict health outcomes, and the accompanying controversy with regard to the possible impact (both helpful and negative) of this information on people's lives. As a breast cancer survivor, and knowing that the genetic alteration that increases the risk of cancer is common to my Jewish ancestry, I have so far chosen not to be tested to see if I have inherited the altered gene. I struggle with whether the uncertain benefits of knowing my genetic status are worth the difficult options such knowledge may suggest, and as information about genetics and disease increases, similar challenges will face many people with regard to many different health situations.

Debra Seltzer  
Administrator, Violence and Injury Prevention Program

Genes may play a role in the risk of many of the most common causes of sickness and death among women, including cancer, cardiovascular disease and diabetes. The most reliable way to identify those at risk for an inherited susceptibility to chronic disease is through their family health histories.

Breast cancer affects 1 in 8 women over their lifetime, and colon cancer affects 1 in 15 women. Approximately 10 percent of breast, ovarian and colon cancer cases are due to inherited mutations in specific genes that can be passed down from either parent and greatly increase the risk of cancer. The genetics of all cancer is complex, and even those individuals in whom single-gene mutations cannot be identified may still have an elevated risk for cancer, emphasizing the importance of knowing one's family history.

Coronary heart disease is the leading cause of death for women in the United States. Although there are significant, modifiable lifestyle risk factors such as smoking, hypertension and obesity, genetics is important in identifying women and men at risk for heart disease and other chronic conditions. Having a male first-degree relative (parent or sibling) who had a heart attack or stroke before age 65, or a female relative who had a heart attack before age 55 is a risk factor for heart disease.

Type 2 diabetes is also a major cause of illness in women. Although obesity and reduced physical activity are the most important risk factors for type 2 diabetes, the more relatives affected with diabetes, the higher the risk to family members.

**Genetic testing is one way to identify the subset of high-risk women who have inherited a susceptibility to cancer.**

In 2005, 1.5 percent of women reported having a genetic test for cancer risk. Among these women, breast cancer risk was most commonly tested (52.9 percent), followed by ovarian cancer risk (38.0 percent) and colon or rectal cancer risk (18.5 percent). Additionally, nearly 24 percent had a genetic test for some other cancer risk. [Respondents could report more than one type of genetic test.]



# Genetics Services in Ohio

By Shelley Nottingham, M.S.W, Genetic Program Coordinator  
Bureau for Children with Medical Handicaps, Ohio Department of Health

## What are genetic services?

Genetics is the branch of medicine that looks at how a family's genes play a role in causing a disease or birth defect. Some people have increased chances for health problems such as cancer or heart disease because of their family's genes. Almost all disease is a result of the interaction between our genes and the environment.

Concerns about health or development may arise at any age. When problems do occur, family members often wonder about:

- The cause
- The impact on one's life
- Other related health issues
- Risks to family members
- Where to find information
- Where to find support

Genetic services may help answer some of these questions. Genetic counseling is a health service that provides families with information and support on a variety of conditions. These conditions include developmental delay, birth defects, genetic disorders and adult-onset chronic diseases. A genetic evaluation may be done to find a reason for a family member's physical difference, developmental delay or health problem.

## Will I benefit from genetic services?

You may benefit from genetic services if you are looking for information about, or support for, a medical condition with a known or possible genetic component affecting you or a close family member. For example:

If you have questions about conditions or traits that run in your family.

- If you have family members with birth defects like cleft palate or genetic disorders like Down syndrome, Huntington's disease or Muscular dystrophy.
- If you share similarities with any of the following groups of people:
  - **Women** whose pregnancy may be at increased risk for complications or birth defects based on age, blood tests, ultrasound findings or family history.



- **Women** who have had two or more miscarriages, a stillbirth or a baby who died.
- **Women** considering pregnancy or who are pregnant and taking prescription medications.
- **Couples** planning a pregnancy who would like more information about genetic conditions that occur more frequently in their ethnic group.
- **Couples** who would like prenatal testing.
- **Couples** who are blood relatives.
- **Parents** whose child has any of the following: developmental delays, a birth defect, behavioral problems, health issues in multiple body systems or has been diagnosed with a genetic condition.
- **Individuals** who are concerned about their personal risk for disease based on lifestyle, occupational exposures, medical history or family history.
- **Individuals** who are concerned about their risk of developing cancer based on personal or family history.
- **Individuals** with a genetic condition who want to learn more about the cause, inheritance, features and possible testing or treatment options.
- **Adopted individuals** now planning a pregnancy

who want to learn about populations risks and prenatal screening.

### **When should I consider seeking genetic services?**

**Before becoming pregnant** – preconception services are available if you are interested in learning about your chances of having a baby with a birth defect or genetic condition before becoming pregnant. Genetic counselors can assess risk and discuss ways to improve the likelihood of having a healthy baby.

**During pregnancy** – advances in technology have led to improved prenatal screening and diagnostic testing options for genetic conditions and birth defects. Genetic counselors obtain family history and pregnancy histories, provide accurate risk assessment, explain prenatal tests and treatment alternatives and provide supportive counseling. Information is delivered in a non-directive manner so you can make your own decisions when contemplating prenatal testing.

**When your child is born** – all birthing centers in the United States participate in newborn screening. Newborn screening is the practice of testing every newborn to detect harmful or potentially fatal disorders that aren't otherwise apparent at birth. Many of these are metabolic disorders that interfere with the body's use of nutrients to maintain healthy tissues and produce energy. Other disorders that may be detected through newborn screening include problems with hormones or blood. A positive newborn screen does not necessarily mean a child has a genetic disorder. When a child has a positive screen, the parents and the child's doctor are notified so further testing can be done to determine if the child has a genetic disorder.



**During childhood** – you may want to seek genetic services if your child has: a known or suspected genetic condition; one or more birth defects; unexplained developmental delay, cognitive impairment or mental retardation; physical, social, emotional or learning problems; or a type of cancer having a suspected hereditary component.

Identifying a cause of your child’s developmental problems can be helpful. Having a diagnosis may affect your child’s education plan, allow for identification of associated health problems, explain your child’s behavior or appearance and inform you of recurrence risks with siblings or future children.

**During adulthood** – some genetic conditions may not become evident until adulthood. It also is becoming clear that a number of common adult-onset conditions, such as cardiovascular disease and diabetes, may have hereditary components. Information, risk assessment, genetic testing and support are available for affected adults as well as those at risk for adult-onset genetic conditions such as Huntington’s disease, hereditary cancers and polycystic kidney disease.

#### **Why are genetic services important?**

Genetic services are important and valuable for a variety of reasons. A genetic consultation can:  
 Help clarify genetic risks.  
 Provide a diagnosis for unexplained symptoms.

Provide information for family members.  
 Identify an unrecognized need for specialized care.

Help you and your family gain access to new testing, clinical trials and support organizations.

#### **What Genetic Services are available in Ohio?**

The Ohio Department of Health (ODH) Genetics program funds a regional network of genetic centers that provides comprehensive care and services to people affected with, or at risk for, genetic-related disorders. The goals of the Regional Comprehensive Genetic Centers (RCGCs) are to assure that:

- 1.** Children and adults with or at risk for birth defects or genetic disorders, and their families, receive quality, comprehensive, genetic services that are available, accessible and culturally sensitive; and
- 2.** The general public and professionals/providers are aware and knowledgeable about birth defects, genetic conditions and genetic disease-related services in Ohio.

For a list of RCGC locations in Ohio, go to <http://www.odh.ohio.gov/odhPrograms/cmh/generv/generv1.aspx> .

Click on **Publications** and then:

**Program Brochure: The Regional Comprehensive Genetic Program.**



# Preparing for a Healthy Pregnancy

By **Connie S. Motter MS, CGC, Genetics Center** Children's Hospital Medical Center of Akron



Every woman wants to have a healthy baby and there are many factors, both genetic and environmental, that can affect a pregnancy. At first glance, these two factors may appear to be unrelated when, in reality, they are often intertwined. Although we cannot change our genes, knowing about certain genetic conditions ahead of time can provide us with the knowledge to make choices

regarding childbearing and optimize pregnancy outcome when our genes place us at an increased risk.

We have even more control over influencing pregnancy outcome by modifying environmental risk factors.

Whether we are talking about genetic or environmental factors that can impact pregnancy outcome, the most important message to take away is that **preparation** is the most effective way to ensure the best possible chances of having a healthy baby.

Historically, preparation for a healthy baby begins at the first prenatal visit. However, we now know this is too late, because the outcome for a baby is set into motion in the womb before a woman ever sets foot into her obstetrician's office. By the time she makes her first appointment, her baby's heart is beating and the spinal cord has formed. So, the best time to plan for a pregnancy is

BEFORE a pregnancy occurs. Here are some important steps to take when planning a pregnancy:

#### Family History

- Know your and your partner's family history. Certain conditions or birth defects may place you or your baby at a higher risk, or you may have a higher risk based on your age or ethnic background. A genetic counselor is best equipped to address family history concerns.

#### Pregnancy History

- Has it been at least 18 months since you had your previous child? This is optimal baby spacing to reduce the risk of prematurity and other adverse pregnancy outcomes.

- If you have had prior miscarriages, pregnancy complications or an adverse pregnancy outcome, review these with your physician prior to getting pregnant, so steps can be taken to minimize future risks.

#### Maternal Health

- Make sure you keep all recommended physician and dental appointments. A preconception visit with your doctor to discuss how to prepare for a healthy pregnancy is optimal.

- Talk with your physician about any over-the-counter medications, prescriptions or herbal supplements to make sure these are safe to take during pregnancy. Some prescription medications that are especially concerning are Accutane, Warfarin, ACE inhibitors and statins.

- Talk with your doctor about health conditions you may have that put you and your

baby at a higher risk, such as hypertension, obesity, diabetes, hypothyroidism, heart conditions, clotting disorders, lupus, PKU and epilepsy. Some medications used to treat your condition may be harmful to your baby, and safer alternatives may be available.

- Talk with your doctor about your immunity to certain infections, such as chickenpox, and whether you need immunizations that can be life saving for you and your baby. Learn how to prevent infections, including those that are sexually transmitted, and ways to treat them.





### Maternal Nutrition

- Know what your healthy weight should be and try to achieve this before the pregnancy. Women who are obese or underweight are at increased risk for having babies with birth defects or who are born prematurely. We are what we eat, and what we put in our bodies is what fuels our bodies, feeds our cells and influences our baby's prenatal growth. This includes eating a balanced diet of whole grains, vegetables, fruits and lean meats. Regular exercise will help maintain a healthy weight and make you feel better.
- Make sure all meat, including lunch meat, is cooked thoroughly to avoid infections such as toxoplasmosis, that can cause birth defects.
- Know the restrictions on the amount of tuna and locally caught fish you should eat. Exposure to mercury can impair fetal development.
- Minimize caffeine consumption to 200 milligrams (mg) a day (which is equal to about one 12-ounce cup of coffee).
- Take a multivitamin containing 0.4 mg of folic acid **every day**. Folic acid helps to prevent birth defects in brain and spine formation. Taking a multivitamin helps ensure you get all of the nutrients that may be missing from your diet.

### Lifestyle

- Assess your social situation to determine if this is the right time to have a baby. Physical and mental abuse, dangerous living situations, lack of partner and/or family support, financial instability and stress can affect pregnancy outcome.
- Do you have medical insurance? Do you have easy access to medical care and other services?
- How is your mental health and stability? Are you experiencing stress, anxiety or depression? Do you have positive outlets for stress and relaxation built into your life? If you don't, seek help from your physician and/or mental health professional.
- If you smoke, drink alcohol or use illegal drugs – STOP! If you can't, you need to get help from your physician and/or a substance abuse professional.

### Environmental Exposures

- Be aware and ask about potentially dangerous exposures at home or work such as lead, pesticides and solvents.
- If you have a cat or rodent, make alternative arrangements for someone to change their cages.

### Additional Resources:

<http://www.marchofdimes.com>

<http://www.cdc.gov/ncbddd/preconception/default.htm>

<http://www.odh.ohio.gov/ASSETS/E71F980AD2F04622A9687B4D1D0BBA90/Birth%20Defects%20Handbook.pdf>

# TESTING FOR HEREDITARY BREAST AND OVARIAN CANCER: QUESTIONS AND ANSWERS

This article has been excerpted with permission from The Ohio State University Clinical Cancer Genetics Program Fact Sheet

By Heather Hampel, MS, CGC, Clinical Associate Professor, Genetic Counselor  
The Ohio State University, Division of Human Genetics

## Q How common are breast and ovarian cancer?

**A** Breast cancer is a common cancer among American women, affecting about 1 in 10 during their lifetime. More than 200,000 women are diagnosed with breast cancer each year in the United States. Breast cancer can also occur in men, but is rare, with fewer than 2,000 cases diagnosed each year in the United States.

Ovarian cancer is not as common as breast cancer, with around 22,000 cases diagnosed per year in the United States and it does not occur in men. One in 70 women in the general population will develop ovarian cancer during their lifetime.

## Q What is hereditary breast and ovarian cancer syndrome?

**A** It has been estimated that about 5 to 10 percent of all breast and ovarian cancer cases are hereditary; in other words, they occur as the result of an inherited predisposition. The most common hereditary breast and ovarian cancer condition is hereditary breast and ovarian cancer syndrome. This means the individual has inherited an increased likelihood for developing breast and ovarian cancer. In most families with an inherited predisposition, several individuals will be affected with cancer. It is important to remember; how-

ever, that the majority of breast and ovarian cancer is *not* hereditary. In fact, breast cancer is so common that it is possible for a family to have several affected women due to chance alone.

## Q How can hereditary breast cancer syndrome be inherited?

**A** BRCA1 and BRCA2 are genes that control cell growth and division. If there is a mutation within either a BRCA1 or a BRCA2 gene, it can no longer control cell growth and division. Cancer occurs when cells grow in an uncontrolled way. Therefore, an altered BRCA1 or BRCA2 gene increases the likelihood that cancer will develop. For this reason, individuals with mutations in either of these genes tend to be diagnosed with cancer at an earlier age and are more likely to develop breast cancer in both breasts (bilateral cancer). BRCA1 appears to be responsible for disease in 45 percent of families with multiple cases of breast cancer only, and in up to 90 percent of families with both breast and ovarian cancer. Studies have shown that there are two specific alterations in the BRCA1 gene which are more common in Jewish individuals of Eastern European ancestry (Ashkenazi Jews).



## Q Is it possible to receive an unclear BRCA test result?

**A** Approximately 10 percent of individuals who undergo complete gene testing for both the *BRCA1* and the *BRCA2* gene, are found to have an unclear result. This means that the gene test was neither positive nor negative. Unclear results occur when an individual is found to have a certain type of gene alteration that may or may not cause an increased chance for developing cancer. In some cases, it may be possible to determine whether the gene change is associated with an increased risk for cancer with additional testing. In other cases, it may take years before we know the meaning of the result.

## Q What if testing reveals that an individual has inherited an altered BRCA1 gene?

**A** Women who inherit a *BRCA1* mutation have an increased chance for developing breast and ovarian cancers. Research is taking place to determine the exact risks of developing these cancers. These risks may vary from family to family and from one individual to another. Women who have an altered *BRCA1* gene have a 56 to 85 percent risk of developing breast cancer by age 70. Their risk for ovarian cancer is 39 to 46 percent by age 70. A woman who has already had



breast cancer and is found to have an altered *BRCA1* gene has an increased risk for breast cancer in her other breast.

## Q What if testing reveals that an individual has inherited a BRCA2 mutation?

**A** Both men and women who inherit a *BRCA2* mutation have an increased chance for developing breast cancer. Women who have an altered *BRCA2* gene seem to have a similar risk of developing breast cancer as found with *BRCA1*. Women with *BRCA2* mutations have a 10 to 20 percent risk for developing ovarian cancer by age 70. Individuals with *BRCA2* mutations seem to also have slightly increased risk for developing pancreatic cancer.

## Q If a woman has an altered BRCA gene, what are the options?

**A** Women with an altered *BRCA* gene have several choices. They can utilize screening methods to help find cancers at the earliest possible stage when they are most easily treated. Women with *BRCA* alterations should make sure to practice breast cancer screening. This means having yearly breast imaging (by mammography and MRI), having breast examinations by a physician and performing monthly breast self-examinations. Screening for ovarian cancer is more difficult. However, women may participate in research that is looking at ultrasound to see if this test, together with pelvic examinations and certain blood tests, can help find ovarian cancer early.

Another option is to take medication to reduce the risk of cancer, called chemoprevention. Both Tamoxifen and Raloxifene (Evista) have been shown to decrease the risk for breast cancer in high-risk, postmenopausal women. Oral contraceptives (birth control pills) have been shown in some studies to decrease the risk for ovarian cancer; however, other studies have shown no benefit.

A third option is to have surgery to remove at-risk body parts before cancer develops. An example of this would be having risk-reducing mastectomy; surgery to remove the breasts to try to prevent breast cancer. Women can also have their ovaries and fallopian tubes removed to try to prevent ovarian cancer. This is called risk-reducing bilateral salpingo-oophorectomy. If this surgery is per-

formed before age 50, it also decreases the risk for breast cancer in women with *BRCA* mutations. Because ovarian cancer screening has not been proven to be effective, we recommend that women with *BRCA* mutations have risk-reducing salpingo-oophorectomy when they have completed childbearing.

## Q What are the possible risks and benefits of *BRCA1* and *BRCA2* testing?

A The only physical risk of testing is that of a routine blood draw. However, other risks and benefits should be considered before undergoing testing. The process of genetic testing may be emotionally difficult whether or not a *BRCA* gene mutation is found. Finding a mutation may indirectly provide information about other family members, who may have chosen not to be tested. In addition, costs for the cancer screening and prevention options may or may not be covered by health insurance. Another issue of genetic testing is the possibility that the results could be used by an employer or insurance company to discriminate against a person. The Genetic Information Nondiscrimination Act (GINA) was signed into law in May 2008. GINA makes it illegal for health insurers to deny insurance coverage or charge a higher rate or premium to an otherwise healthy individual found to have a potential genetic condition or genetic predisposition toward a disease or disorder. Protections in health insurance went into effect either in May 2009 or when a large group policy first renews after that date. GINA also makes it illegal for employers to use an employee's genetic information when making hiring, firing, placement or promotion decisions. Employment protections were being implemented in November 2009. Currently, the ability to obtain life and disability insurance is not protected by any laws.

The decision to participate in *BRCA1* or *BRCA2* testing is a complicated one. Individuals and families must weigh the risks and benefits of testing; they must also consider their unique situations. Ultimately, individuals must make their own decisions.

To locate cancer risk assessment services in your region, go to:  
<http://www.odh.ohio.gov/odhPrograms/cmh/genserv/genserv1.aspx>.

# ORGANIZATIONS AND ASSOCIATIONS

## Genetic Alliance

4301 Connecticut Avenue NW - Suite 404  
Washington, DC 20008-2369  
Phone: 202.966.5557 Fax: 202.966.8553  
[info@geneticalliance.org](mailto:info@geneticalliance.org)

## About Us

Founded in 1986 as the Alliance for Genetic Support Groups, Genetic Alliance has become the world's leading nonprofit health advocacy organization committed to transforming health through genetics. Our open network connects members of parent and family groups, community organizations, disease-specific advocacy organizations, professional societies, educational institutions, corporations and government agencies to create novel partnerships. We actively engage in improving access to information for individuals, families and communities, while supporting the translation of research into services.

## Mission

We promote an environment of openness centered on the health of individuals, families and communities.

## Goals

Our goal is to build capacity within the genetics community by being fluid, dynamic, and efficient. We work to eliminate obstacles and limitations through partnerships, informed decision making and identifying solutions.

## The Genetics and Public Policy Center (GPPC)

Johns Hopkins University  
Berman Institute of Bioethics  
1717 Massachusetts Avenue, NW, Suite 530, Washington, DC 20036  
Phone: 202.663.5971,  
Fax: 202.663.5992

## The GPPC Vision

The GPPC was created in 2002 at Johns Hopkins University by Pew Charitable Trusts to help policymakers, the press and the public understand and respond to the challenges and opportunities of genetic medicine and its potential to transform global public health.

## The GPPC Approach

The GPPC conducts rigorous legal research and policy analysis, performs policy-relevant social science research,

crafts robust policy options and recommendations and convenes and consults key stakeholders to identify common ground and develop consensus and influences national genetics programs and policy.

In order to realize the potential of genetic medicine, the GPPC has a detailed plan to:

Develop and advocate for policies ensuring the safety and effectiveness of genetic tests and treatments.

Create models for fair systems of reimbursement to ensure broad access to genetic medicine and incentives for innovation.

Shift the current paradigm for creating, disseminating and adopting medical and industry guidelines to a highly responsive and evidence-based system that benefits the public.

Protect genetic information so the public feels safe taking a genetic test and participating in genetic research.

Build public understanding of, and trust in, genetic medicine.

## MARCH OF DIMES

The March of Dimes Making A Difference

Our mission is to improve the health of babies by preventing birth defects, premature birth, and infant mortality. The March of Dimes carries out this mission through programs of research community services, education and advocacy to save babies' lives. The March of Dimes is working to determine what causes premature births and conduct genetic research to find cures for genetic birth defects.

The March of Dimes helps pregnant women know what to worry about and what not to worry about when it comes to having a healthy baby. Through our [Pregnancy and Newborn Health Education Center<sup>SM</sup>](#), women can get free, one-on-one, confidential answers to their questions about pregnancy, preconception, newborn screening and related topics.

The March of Dimes also provides a wide variety of materials including *mama*, an annual magazine full of practical and important information for parents-to-be, are available. Go to: <http://www.marhofdimes.com> to find your local chapter.

## Ohio Department of Health, Violence and Injury Prevention Program

– Debra Seltzer, Administrator

### SEXUAL ASSAULT AND DOMESTIC VIOLENCE PREVENTION PROGRAM:

Debra Seltzer, Program Administrator (614) 728-2176 – [Debra.Seltzer@odh.ohio.gov](mailto:Debra.Seltzer@odh.ohio.gov)

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Women's Health Program Web Site: [http://www.odh.ohio.gov/pdhPrograms/hpr/wom\\_hlt/sadvhlth.aspx](http://www.odh.ohio.gov/pdhPrograms/hpr/wom_hlt/sadvhlth.aspx)