



## “What’s in Your Genes?”: Women’s Health & the Role of Genetics

“What’s in your genes?” [1] It’s a simple question, but one that doctors have spent decades trying to answer.

Our genes shape who we are. From our hair and eye color to our height, we see in us our mothers and fathers, our grandmothers and grandfathers.

But genetics also play an important role in our health as women.

When visiting a new doctor, you’ve likely been asked, “Do you have a family history of breast cancer?” Ovarian, cervical, and other reproductive cancers might also be on that list.

Roughly 12% of women in the U.S. are diagnosed each year with breast cancer – and 5-10% of those women develop breast cancer due to an inherited gene mutation.[2]

Precision Imaging Centers powered by HALO Diagnostics cares for many women with family histories of cancer. Patients may come to see us for services such as a routine mammogram, a diagnostic screening after developing breast cancer before menopause,[3] or for a second opinion.

Annual breast screenings and genetic testing can work together to create targeted therapies, offer early detection, and improve women’s health.

### Your Genes & Disease

Let’s start from the top. How do our genes predispose us to disease?

We’re not that different – 99.9% of someone’s genes are the same as yours, your mother’s, your sister’s, and so on. Utterly and identically human.

But the remaining 0.1% of our genes makes all the difference. That genetic makeup is a wildcard: We might inherit a mutation from one of our parents that increases our risk for obesity, heart disease, cancer, or diabetes.

Still, simply having this variant doesn’t automatically lead to disease. Its impact may be large or small. Your environment, lifestyle, and even other genetic factors may play a role.[4]

### Women’s Health

Breast exams. Pap smears. Bone density tests.

These tests are just some (of many) that help keep your health on track. No doubt about it – your health is complex, and that includes your genetics.

You’re probably familiar with a few gene variants affecting women. For example, having *BRCA1* and *BRCA2* mutations notably increases your risk for breast and ovarian cancers. Changes in *BARD1* and *BRIPI* genes pose a smaller breast cancer risk.[4] There are also mutations in the *MLH1* gene, which can contribute to the development of uterine and colorectal cancers.[5]

### Ins & Outs of Genetic Testing

Genetic testing is a deeply personal choice. Don’t fret if you have doubts or questions – there are steps you can take to ensure you’re making the right decision for you and your health.

#### Pre-test Counseling

You need answers to your questions.

Common concerns include, “What test is right for me?”, “What happens if I have a mutation?”, and “Are my family members at risk for the same?”

We suggest scheduling some time with your healthcare provider – your primary care physician, gynecologist, or breast specialist – to talk about the pros and cons of genetic testing. Before going forward, your doctor will discuss what test(s) you may need, the benefits and limitations, and what to expect next.

Sometimes, your doctor won’t have all the answers. They may refer you to a genetic counselor, gynecologic oncologist, or other specialist.[6]

#### Types of Genetic Tests

Together, you and your doctor will decide what test(s) you’ll receive. Genetic tests for women’s health include (but aren’t limited to):

- Pre-symptomatic testing helps to estimate your risk of developing a disease such as breast or ovarian cancer.
- Prenatal screenings identify the likelihood of your baby having birth defects, which may be due to genetic disorders.
- Newborn screenings check for genetic disorders at birth that may be rare and have the potential to cause major health issues.
- Carrier screenings let you know if you’re a carrier for a certain genetic condition.[7]

#### Taking the Test

It’s finally arrived: genetic testing day. After checking in, you’ll meet with your doctor (or a member of their team such as a nurse or phlebotomist) to collect your samples. You’ll be asked to provide one or more samples of blood, skin, saliva, or fluid to send to the lab for testing.[8]

#### At the Lab

Now, your genes’ real journey begins.

Your DNA samples go through a four-step process: preparation, sequencing, analyzing, and interpreting. The lab may look at your DNA in a variety of ways, with the goal of identifying any variants that cause or increase your risk of disease.

Genetic counselors, doctors, scientists, and even computer experts come together to review and interpret your results.[8]

#### Getting Your Results

Your doctor will receive a written copy of your test results. Next, they’ll contact you to discuss your results and determine next steps.

Let’s consider this scenario: If your test(s) show mutations in the *BRCA1*, *BRCA2*, *BARD1*, or *BRIPI* genes, then additional tests around breast and ovarian cancers, as well as close monitoring and regular mammograms, may be needed[9]. Every woman’s level of risk is different. So is the treatment.

Whether you’re still uncertain, simply researching, or actively pursuing tests, genetic testing is your journey to take. Genetics open an important window into women’s health – and offer the possibility of a better future.

## Contact Us

Precision Imaging Centers powered by HALO Diagnostics offers life-saving breast screenings and diagnostic tests at our four Florida locations in Jacksonville, Jacksonville Beach, Fleming Island, and St. Augustine.

Schedule your mammogram today. Most insurers cover annual mammograms. We make it easy, and it takes 15 minutes or less. Give us a call at (904) 996-8100. You can also schedule your mammogram online.

[1] McCormick, K. (2015). Genetic testing and its role in women’s health and cancer screening. *Stanford Medicine*. Retrieved from <https://scopeblog.stanford.edu/2015/07/28/qa-whats-the-role-of-cancer-genetics-in-womens-health/>

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