FANCONI™ CANCER FOUNDATION

Frequently Asked Questions: Genetic Counseling for FA

WHAT IS A GENETIC COUNSELOR AND HOW DO I GET ONE?

Genetic counselors are healthcare professionals with specialized training in medical genetics and counseling. They help people understand how inherited conditions like Fanconi anemia (FA) may affect them or their families. They also explain genetic test results in the context of your personal and family history.

Genetic counselors work in many areas of healthcare, including:

- Reproductive and prenatal care
- Pediatrics
- Cancer and oncology
- Neurology
- And other specialties

They're a valuable part of your care team when genetic information is part of the picture.

The National Society of Genetic Counselors (NSGC) has a directory that can be used to find a genetic counselor in your area.

QUESTIONS OR NEED SUPPORT?

1-888-326-2664 | 541-687-4658
communitysupport@fanconi.org
fanconi.org

HOW DOES ONE GET FA? WHAT'S THE DIFFERENCE BETWEEN A CARRIER AND SOMEONE WITH FA?

FA is usually inherited in a recessive way, which means **a person must inherit two non-working copies of the same FA gene (one from each parent) to have FA**. A small number of FA cases (specifically those caused by changes in the FANCB gene) are inherited in an X-linked pattern. In these cases, only one copy of the defective gene is disease-causing.

People with only one non-working copy are called carriers. Carriers don't typically have symptoms or health problems from FA. However, for a few specific genes (like *BRCA1, BRCA2, BRIP1, PALB2, RAD51C*, and *RAD51D*), being a carrier can increase a person's cancer risk.

If both parents are carriers of the same FA gene, there's a 25% chance with each pregnancy that their child will inherit both non-working copies and have FA.

We're still learning more about what it means to be a carrier for other FA-related genes. If you're found to be a carrier, a genetic counselor can help you understand what that might mean for your own health and your family's future. They can also guide you through testing options for your partner and explain the chances of having a child with FA. This can be done before or during pregnancy.

Continued on following pages

UNDERSTANDING TEST RESULTS

Think of genes like very long words, each with a specific spelling. Genetic testing is like running spell-check. It looks for changes in how those words are spelled.

Some spelling changes are harmful and affect how the gene works. These are called **pathogenic or likely pathogenic variants**. You might also hear the term "mutation."

Some changes are just normal differences between people. These are called **benign or likely benign variants**, and they don't cause problems. Most people have many of these, and test reports often don't mention them.

Sometimes, the test finds a change but it's unclear if it's harmful or not. This is called a **variant of uncertain significance** (VUS). It just means we need more information. Over time, as researchers learn more, the lab may reclassify it as either benign or pathogenic.

If you receive a genetic test result with a VUS or other unfamiliar term, a genetic counselor can help you understand what it means and what to expect next.

WHEN WOULD SOMEONE SEE A GENETIC COUNSELOR MORE THAN ONCE?

In most cases, genetic counseling is a one-time consultation. For example, if you're found to be a carrier for FA, one visit with a genetic counselor is usually enough to explain your results and what they might mean for your health and family.

Ongoing follow-up with a genetic counselor isn't typically needed in these situations.

However, if someone is diagnosed with FA, they may have long-term care involving many healthcare specialists. This may include regular visits with a geneticist or genetic counselor as part of their care team.

WHAT TO EXPECT WHEN MEETING WITH A GENETIC COUNSELOR

When you meet with a genetic counselor, they'll ask about your personal health history and your family's medical history. It's helpful to come prepared to talk about your relatives—especially parents, siblings, grandparents, aunts, uncles, and cousins.

If anyone in your family has had major health problems or a known genetic condition, your counselor will use that information to better understand your risks and guide testing or next steps.

You may be wondering what types of questions to ask. Here are some suggestions:

- Does being a carrier of FA impact my healthcare based on the specific genetic result I have?
- Should my partner be tested to see if they are a carrier for FA?
- If I am planning a family with an egg or sperm donor, should the egg or sperm be tested?
- Should my current children be tested to see if they have FA?

DOES INSURANCE COVER GENETIC COUNSELING OR TESTING?

Insurance coverage varies depending on your health plan. It's a good idea to check with your insurance provider to see what's covered and ask your primary care provider for a referral, which may be required.

In most cases, genetic counseling and genetic testing are billed separately. A genetic counselor can help you understand what your insurance might cover.

Many genetic testing labs also offer patient support programs, including financial assistance, lower-cost testing, and sometimes no-cost testing, depending on your situation.

WHAT KINDS OF GENETIC TESTS ARE USED FOR FA?

The definitive test for diagnosing Fanconi anemia is a **chromosome breakage test**. This test looks at how a person's blood cells respond to certain chemicals that damage DNA. In people with FA, the cells can't repair the damage well, leading to visible chromosome breaks under a microscope.

- The chemicals most commonly used are DEB (diepoxybutane) and MMC (mitomycin C).
- These tests can also be done during pregnancy using cells from chorionic villi or amniotic fluid.

WHEN IS FA TESTING RECOMMENDED?

- If a baby is born with thumb, arm, or other physical differences linked to FA
- If a person develops aplastic anemia (a bone marrow failure disorder) at any age—even if they have no other symptoms
- Before bone marrow transplant for aplastic anemia, since people with FA need different treatment due to sensitivity to radiation and chemotherapy

Some people with FA do not have visible physical signs, so testing is essential for an accurate diagnosis and proper medical care.

NEED HELP FINDING THE RIGHT RESOURCE?

If you're looking for expert care or guidance, you can also reach out directly to one of the FA Clinical Care Centers for more information. These centers specialize in the diagnosis and care of individuals with FA.

You're also welcome to contact the Fanconi Cancer Foundation (FCF) team at any time—we're here to support you and help you navigate your options.

GENETIC TESTING AND COUNSELING RESOURCES

Before making an appointment, it's important to contact your health insurance provider to confirm whether the clinic or provider is in-network, and what your co-pay, co-insurance, or deductible may be.

If the provider is out-of-network, you may still be able to use their services. Your insurance company can help you explore options, including whether FA-related testing and counseling can be done at a specialized center or within your plan's network.

Find a Genetic Counselor Tool

Use this tool from the National Society of Genetic Counselors (NSGC) to search by location, type of appointment (in-person or telehealth), and specialty (Adult, Pediatric, Cancer, Hematology, Prenatal, and others).

Grey Genetics (Telehealth Counseling)

Grey Genetics offers online genetic counseling through a nationwide network of licensed counselors.

- Self-pay only (they don't accept insurance), but fees are reasonable
- They provide a receipt for possible insurance reimbursement
- Self-referrals are welcome—no doctor's referral needed

Recommended Options:

- For those affected by or at risk for Fanconi anemia, book the "Adult Genetics – Other" consult for an in-depth discussion
- For general questions, a free 10-minute consult is also available

Questions? Contact them at:
info@greygenetics.com