What is the best way to find a genetic counselor?

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

How would you explain FA to a family in lay terms? What's the difference between a carrier and someone with FA?

Fanconi anemia is primarily a recessive disorder, which means that both copies of a gene are defective (not working correctly) for a person to have FA. This happens when 2 parents are CARRIERS, meaning each parent CARRIES 1 defective copy of an FA gene, but they have 2nd copy of the gene that is working like it should. If two parents are both carriers of the same defective FA gene, there is a 25% chance that a child will inherit BOTH defective copies of the gene, one from each parent.

Being a carrier (1 defective copy of an FA gene) usually does not cause medical problems.

However, there are SOME genes when being a carrier does have medical impact for parents. (BRCA1, BRCA2, BRIP1, PALB2, RAD51C, RAD51D).

We are learning more about what it means to be a carrier for other FA genes (not the ones listed above). It is possible that we could know more in the future.

If you are found to be a CARRIER for FA, it would be helpful to talk to a genetic counselor to learn more about the specific gene you are a carrier of, and if it has any impact for you.

If you are planning a family, genetic counselors can also help explain the risk for FA in any future children and if your partner needs carrier screening as well. This can be done before or during pregnancy.

In what instances would someone see a genetic counselor multiple times for follow-up?

Most often, genetic counseling is a consultative service, meaning that most individuals usually meet a genetic counselor for one consultation but do not have long-term follow up.

For example, if you are found to be a carrier for FA, you can likely meet with a genetic counselor for one visit to understand your results and impact of the results for you and your family. You will most likely not need long-term follow up with a genetic counselor in this case.

Children and adults diagnosed with FA have long-term follow up with different types of healthcare providers, which may include a geneticist or a genetic counselor.

Understanding test results

Genes are like very long words, each with their own spelling. Genetic testing is like a "spell-check" on your genes. Some spelling changes in our genes are problematic, and some spelling changes are simply normal differences between two people.

Pathogenic variants or likely pathogenic variants are spelling errors that are problematic and affect how the gene works (sometimes you might hear the term "mutation" used).

Benign or likely benign variants are spelling changes that are not problematic and determined to be normal differences. Our DNA is slightly different from one person to the next so these benign spelling changes can be common, and genetic test reports/results usually do not comment on them.

Sometimes genetic testing can identify a spelling change, but the laboratory doesn't have enough information to understand if that spelling change is harmful or not. This type of change is called a variant of uncertain significance. The laboratory needs to keep studying these uncertain variants, and over time they may be able to update it to either benign or pathogenic.

Questions to ask a genetic counselor

If you are meeting a genetic counselor after you are found to be a carrier of FA, 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?

When to see a genetic counselor

Who are genetic counselors? Based on the National Society of Genetic Counselors' website:

"Genetic counselors have advanced training in medical genetics and counseling to guide and support patients seeking more information about how inherited diseases and conditions might affect them or their families, and to interpret genetic test results based on your personal and family history."

Genetic counselors can work in different healthcare specialties, such as prenatal/reproductive, pediatrics, oncology, neurology, and many others.

Do most insurances pay for GC? Are there OP costs? Eligibility?

Insurance coverage for genetic counseling can depend on the type of health insurance you have. It's best to check with your own insurance company to see if they will cover a genetic counseling visit and/or if they will cover genetic testing. You can also ask your primary care provider for a referral to a genetic counselor.

Generally, genetic testing is a separate cost from genetic counseling. Genetic counselors help patients determine if their insurance may or may not cover genetic testing. Many genetic testing laboratories

also have various payment options for patients like patient-assistance programs, lower-cost testing, and in some cases no-cost testing.

What to expect when meeting with a genetic counselor

When you meet with a genetic counselor, they will ask you questions about your personal health history and your family history.

It is important to be prepared to tell the genetic counselor about your relatives, including siblings, parents, grandparents, aunts/uncles, and cousins. If there is any family history of major medical problems or genetic diseases, you will share this information with the genetic counselor.

Which types of genetics tests are applicable to FA?

The definitive test for FA is a chromosome breakage test: some of the patient's blood cells are treated, in a test tube, with a chemical that crosslinks DNA. Normal cells can correct most of the damage and are not severely affected, whereas FA cells show marked chromosome breakage. The two chemicals commonly used for this test are DEB (diepoxybutane) and MMC (mitomycin C). These tests can be performed prenatally on cells from chorionic villi or from the amniotic fluid.

Many cases of FA are not diagnosed at all or not diagnosed in a timely manner. FA should be suspected and tested for in any infant born with the thumb and arm abnormalities described previously. Anyone developing aplastic anemia at any age should be tested for FA, even if no other defects are present. Many people with FA show no other abnormalities.

It is essential to test for FA before contemplating bone marrow transplantation for aplastic anemia. The regimen used to prepare patients for transplant is very different for FA patients, as FA patients tolerate radiation and chemotherapy very poorly.

Genetic Testing and Counseling Resources:

Before scheduling an appointment, contact your health plan to verify that the location or provider you plan to visit is included in your network. Your health plan will also be able to inform you of any co-payments, co–insurances, or deductibles that will be your responsibility. If out of network, it may still be possible to receive genetic counseling services at one of these locations. Work with your insurance company to determine whether appropriate FA genetic testing and counseling can be provided at one of these specialized centers or somewhere within your network.

Find A Genetic Counselor Tool: This tool will allow you to search by "in person" or telehealth, and search by city, state and/or specialty. Some specialties include Adult, ART/Preimplantation Genetics, Cancer, Hematology, Pediatric, PGD/Preconception, Specialty Disorders, or possibly prenatal genetic counseling.

Grey Genetics: Telehealth counseling and consulting. They have a network of genetic counselors that can be booked online. They are licensed in nearly every state. Self-referrals are accepted, and their services are self-pay at reasonable rates because they do not accept insurance. They will provide a receipt for insurance reimbursement purposes. For those who may be or are impacted by FA and who

would like to review test results in-depth, they recommend booking the "Adult Genetics Other" consult. For more basic and general information, a free ten minute consult may be booked. If you would like additional information or are unsure of which type of consultation is right for you, feel free to contact them at info@greygenetics.com.

FA Comprehensive Care Centers

Cincinnati Children's Hospital Medical Center
Fanconi Anemia Comprehensive Care Center Bone Marrow Failure Clinic
Cincinnati, OH
Erica L. Goodridge, RN, BSN, CPN, Care Manager
Fanconi Anemia Comprehensive Care Center
513-636-3218
Toll-free: 1-800-344-2462, ext. 3218
erica.goodridge@cchmc.org
https://www.cincinnatichildrens.org/service/f/fanconi-anemia/team

Services:
DEB/MMC diagnostic testing using blood or bone marrow samples.
Complementation analysis for A, B, C, E, F, G, L, I (I analysis via skin testing only) and FANCD2 (via western blot). Testing done with blood, skin biopsy or amniocytes. Note: This testing must be requested by clinicians or researchers.
Molecular testing to find specific mutations for patients with FANCA, FANCC, or FANCG.
Prenatal diagnostic molecular testing for FANCA, FANCC, or FANCG.
Carrier detection testing for patients with a family history of FA, from complementation groups FANCA, FANCC, or FANCG.

Dana-Farber Cancer Institute
The Comprehensive Center for Fanconi Anemia
Dana-Farber Cancer Institute, HIM 208
450 Brookline Avenue
Boston, MA 02215
Phone: 617.632.6302
Fax: 617.632.5757
FA Testing: https://www.dandrealab.org/comprehensive-center-for-fanconi-anemia.html

Alan D'Andrea, MD
617-632-2080
alan_dandrea@dfci.harvard.edu

Lisa A. Moreau MS, CLSpCG
Dana-Farber Cancer Institute, HIM 244
450 Brookline Avenue
Boston, MA 02215
Phone: 617.632.5792

Fax: 617.632.5757
e-mail: lisa_moreau@dfci.harvard.edu
If you have any questions about our diagnostic services, **contact us!**

Services:
DEB/MMC diagnostic testing using blood, bone marrow, or skin fibroblasts.
FANCD2 western blot (Research).
FA carrier testing.

Memorial Sloan Kettering Cancer Center
New York, New York
For U.S. Adult Patients
Call 800-525-2225
Mon-Fri, 8:00 AM - 6:00 PM, ET
For International Patients
Call 212-639-4900

For Child & Teen Patients
Call 833-MSK-KIDS

Genetic Testing and Counseling:
https://www.mskcc.org/cancer-care/risk-assessment-screening/genetic-counseling-and-testing

Elise Fiala, MS
Pediatric Genetic Counselor
fialae@mskcc.org
646-888-4116

FA Comprehensive Care Center:
Maria Cancio, M.D., Hematology
canciom@mskcc.org
Website: https://www.mskcc.org/pediatrics/cancer-care/types/pediatric-blood-disorders/about-pediatric-blood-disorders/inherited-bone-marrow-failure-syndromes

University of Minnesota Masonic Children's Hospital
Center for Pediatric Blood and Marrow Transplantation; FA Comprehensive Care Center
East Building, Ninth Floor
2450 Riverside Avenue
Minneapolis, MN 55454
Telephone: 612-273-2800

Genetic Counseling:
https://www.mhealth.org/childrens/care/services/genetic-counseling

FA Comprehensive Care Center:
https://www.mhealth.org/childrens/care/treatments/blood-and-marrow-transplant-pediatrics

Margaret MacMillan, M.D., Director
macmi002@umn.edu
John Wagner, M.D., Distinguished McKnight Professor
wagne002@umn.edu

FA International Registry:

The Rockefeller University
Part of New York Consortium for Inborn Errors of Hematopoiesis and Immunity (see also, Memorial Sloan Kettering Cancer Center, New York Presbyterian Hospital, and Hospital for Special Surgery)
1230 York Avenue Box 182
New York, NY 10065
Telephone: 212-327-8612
Fax: 212-327-8262
Facebook: *Fanconi Anemia Registry or Agata Smogorzewska* (PI of IFAR)
http://lab.rockefeller.edu/smogorzewska/families/
(no genetic counseling currently available, however, FA testing, diagnosis, and education are provided by an FA expert)

Others Centers that test, treat, and counsel for FA

Lucile Packard Children's Hospital – Stanford
725 Welch Road
Palo Alto, CA, 94304
Outpatient clinic: 1st floor, West building
Inpatient unit: 5th floor, Main building

Bone Marrow Failure Disorders Program:
https://www.stanfordchildrens.org/en/service/bone-marrow-failure-disorders
(650) 497-8953 or email bmf@stanfordchildrens.org.
Agnieszka Czechowicz, M.D., Hematology
aneeshka@stanford.edu
Alice Bertaina, M.D., Hematology
aliceb1@stanford.edu

Genetic Counseling:
https://www.stanfordchildrens.org/en/service/genetics/careteam
(650) 721-5804

Indiana University School of Medicine
Wells Center, Riley Hospital for Children
Indianapolis, IN
Pediatric Genetic Counseling:
https://www.rileychildrens.org/health-info/pediatric-genetic-counseling

Adult FA Testing:
https://geneticslab.medicine.iu.edu/search.html?keywords=fanconi+anemia

https://medicine.iu.edu/genetics/clinical-care

Bone Marrow Failure Care:
https://www.rileychildrens.org/health-info/bone-marrow-failure

Services:
Complementation Group (Research):
Use of retroviruses for A, B, C, D1, D2, E, F, G, I, J, L, M, N, O, P, Q in primary T-cells, bone marrow cells, or fibroblast cells or in lymphoblast cell lines (LCL) (Research).
FANCD2 western blotting (research).
**Individuals should have a primary care physician contact the office at 317-944-3966 to make appropriate referrals and to send medical records.*

Oregon Health & Science University
Knight Cancer Institute - Genetic Counseling Services:
Center for Heatlh & Healing Building 2
3485 S. Bond Ave.
Portland, OR 97239
Patient Phone Line: 503-494-4800
https://www.ohsu.edu/knight-cancer-institute/genetic-counseling-and-risk-assessment

Susan Olson, PhD, FACMG (cytogenics)
503-494-5964
olsonsu@ohsu.edu
https://www.ohsu.edu/people/susan-b-olson-phd

C. Sue Richards, PhD, FACMG
(molecular)
503-494-5400
richarsu@ohsu.edu
https://knightdxlabs.ohsu.edu/home/about-us/medical-directors/director-biography?id=Sue+Richards

Knight Cancer Institute FA Testing Website: https://knightdxlabs.ohsu.edu/home/genetic-disorders/molecular-genetics/fanconi-anemia
Client Services
Phone: (855) 535-1522
Fax: (855) 535-1329
Email: KDLClientServices@ohsu.edu

Services:
Genetic Counseling
DEB/MMC diagnostic testing - blood and skin fibroblasts.
Next-Generation sequencing comprehensive Fanconi panel is now available for all FANC genes. Exon-centric microarray is also available for detection of deletions and duplications in all FANC genes. Complementation and full gene mutation analysis by Sanger sequencing for A, C, G, E, F as well as targeted sequencing of any exon is available. Deletion/ duplication analysis for FANCA is also available

by MLPA. Prenatal testing available for known mutations. Custom sequencing for known mutations in Fanconi genes other than A, C, G, E, F is also available. Please see Gene Tests for further information. FA carrier testing.