

# FAQ

## Frequently Asked Questions

### What is Fanconi anemia?

Fanconi anemia (FA), named for the Swiss pediatrician, Guido Fanconi, is an inherited DNA repair disorder that may lead to bone marrow failure, leukemia, and/or solid tumors. FA can affect all systems of the body. It is a complex and chronic disease that is psychologically demanding. FA is also a cancer-prone disease. Research has added years to the lives of people with FA. Decades ago, children rarely survived to adulthood. Now, there are adults with FA that live into their 30s and beyond.

### What causes Fanconi anemia?

FA is a very rare genetic disorder. FA is primarily a recessive disorder: if both parents carry a defect (mutation) in the same FA gene, each of their children has a 25% chance of inheriting the defective gene from both parents. When this happens, the child will have FA.

### How many FA genes are there?

FA is caused by 22 different genes, including the two breast cancer genes, *BRCA1* and *BRCA2*. The three most common FA genes are *FANCA*, *FANCC*, and *FANCG*.

### Who can have FA?

FA occurs almost equally in males and females and is found in all ethnic groups. The incidence rate, or the likelihood of a child being born with FA, is about 1 in 131,000 in the U.S., with approximately 31 babies born with FA each year in the U.S.

### What are the symptoms of FA?

Individuals affected by FA can experience:

- Birth defects affecting thumbs, forearms, and other parts of the skeleton
- Kidney, urinary tract, and heart malformations
- Digestive difficulties
- Abnormal blood cell counts
- Hearing loss
- Bone marrow failure and/or leukemia, requiring a stem cell transplant
- Certain types of cancers (especially head and neck and gynecologic cancers) at a significantly younger age than the general population, even after a stem cell transplant.
- Intellectual developmental delay

### How is FA treated?

At the present time, stem cell transplantation is the only long-term cure for the blood defects in FA. Stem cells can be taken from a donor's marrow or peripheral blood, or can be obtained through cord blood harvested at the time of a baby's birth. To prepare for transplant, the patient's own bone marrow is destroyed, making space for the new, healthy stem cells to engraft. Donor stem cells can be matched or partially mismatched to the patient's tissue type. The closer the match, the less likely that the new stem cells will recognize the patient's cells as foreign and attack them, a complication known as graft-versus-host disease.

Always consult your physician before taking any action based on the information presented on this page.

### What is the Fanconi Anemia Research Fund?

Lynn and Dave Frohnmayer started the Fanconi Anemia Research Fund, Inc. (FARF), in 1989, to find effective treatments and a cure for Fanconi anemia and to provide education and support services to affected families worldwide. Support includes: medical resource information, education, publications, online support groups, annual family meetings, and meetings for adults with FA.

FARF has awarded over \$20 million to fund more than 225 research grants.

FARF-supported research has made significant improvements in the bone-marrow transplant survival rates of people with FA.

Research funded by FARF has helped to uncover important information about cancers that affect both people with and without FA.

More than 95% of the Fanconi Anemia Research Fund's annual budget comes from family fundraisers.