

## All about

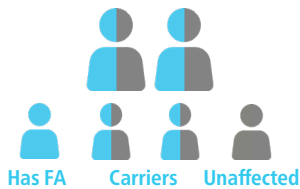
# FANCONI ANEMIA

### What is FA?

Fanconi anemia (FA) is a genetic DNA repair disorder that may lead to bone marrow failure, leukemia, and/or solid tumors (cancer). It is caused by one of at least 23 genes. FA can affect all systems of the body. It is a complex and chronic disease that is psychologically demanding.

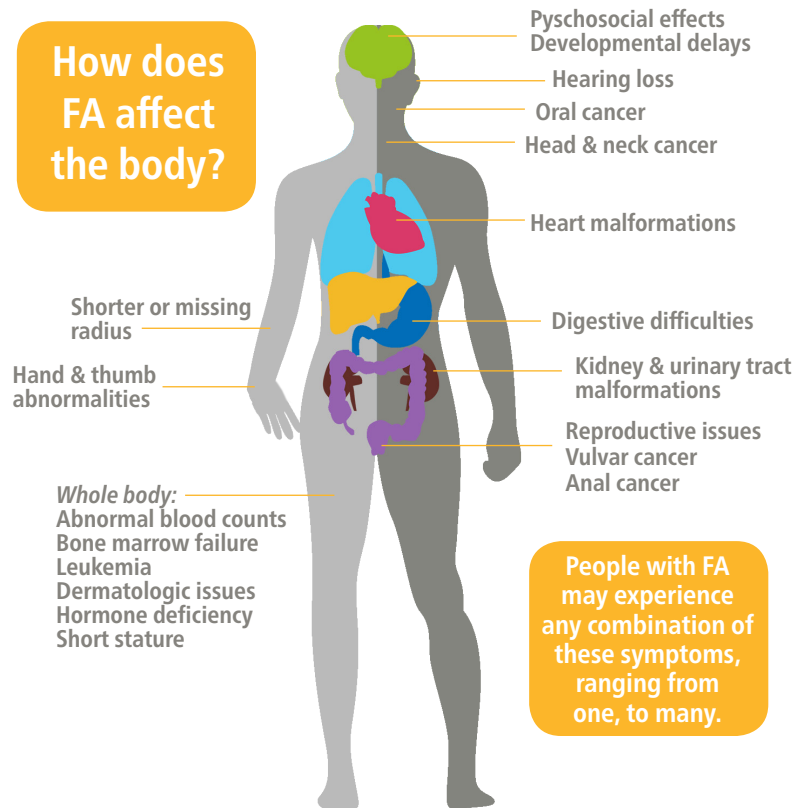
**1 in 131,000**

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



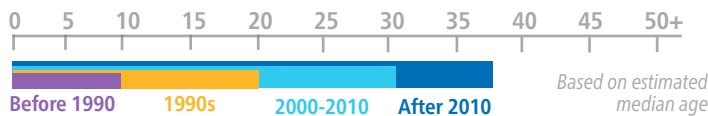
If both parents carry a mutation in the same FA gene, they have a 25% chance of having a child with FA.

### How does FA affect the body?



People with FA may experience any combination of these symptoms, ranging from one, to many.

### Life expectancy



### Here's why you should care

It's simple: children and adults with Fanconi anemia need research to live. Without research, they won't get the advances in treatment that they need to survive. Here's another big reason you should care: FA research benefits the rest of the population, too. Bone marrow transplants have become much safer & more effective because of studies with FA patients. At least five FA genes are also breast cancer susceptibility genes, meaning therapies developed for FA patients would benefit breast cancer patients, too. And, Fanconi anemia research is in the process of unlocking the mysteries of DNA repair problems, which are at the root not only of FA, but of cancer.

### Here's how research helps



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, 40s and beyond.



Thanks to research, the rate of successful bone marrow transplants has gone from 20% in the 1990s to over 90% today.

### Here's what needs to happen next

- ☐ Fund clinical trials aimed at treating and preventing cancers in people with FA
- ☐ Develop treatment plans for all stages of FA cancers, recognizing limitations for those with FA (for example, no radiation)

# We need you.

# 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.