Fanconi Anemia Fact Sheet

FANCONI ANEMIA RESEARCH FUND

WHAT IS FANCONI ANEMIA?

Fanconi anemia (FA) 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. The types and severity of symptoms can differ widely from person to person. Many people with FA undergo hematopoietic stem cell transplants to treat bone marrow failure, precancerous blood cell changes, and leukemia.

Due to the underlying DNA repair problem, people with FA are at a much higher risk of developing a variety of cancers and at a much earlier age than patients in the general population. Those who have had a successful stem cell transplant, and are therefore cured of the blood problem associated with FA, still must have regular examinations to watch for signs of cancer.

Research has resulted in better outcomes and longer lives for those with FA. There is now a growing population of adults living with FA.

WHAT CAUSES FA?

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?

Twenty-three different genes may cause FA, 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 may experience one or several of these symptoms:

- Abnormal blood cell counts
- Birth defects affecting thumbs, forearms, and other parts of the skeleton
- Bone marrow failure and/or leukemia
- Certain types of cancers (especially head and neck and gynecologic cancers)
- Kidney, urinary tract, and heart malformations
- Digestive difficulties
- Hearing loss
- 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. To prepare for transplant, the patient's own bone marrow is destroyed, making space for the new, healthy stem cells to engraft.

Even if the patient has had a successful stem cell transplant, they must still watch for signs of cancer, especially in the head and neck and anogenital regions. Early detection is key. Surgery remains the best course of treatment for those who develop cancers. Surgery may also be used to address other symptoms of FA, like missing thumbs.

THE FANCONI ANEMIA RESEARCH FUND IS HERE FOR YOU

RESEARCH

Our research focuses on cancer, bone marrow failure, FA genes and the DNA repair pathway, and other body systems affected by FA

SUPPORT

We provide 1:1 guidance to people with FA, caregivers and families. We help provide resources needed to manage FA and emotional support for those who want it.

EDUCATION

FA is a complex disease, which is why we create materials to help understand each stage, like our FA Clinical Care Guidelines and topic-specific flyers and brochures.

COMMUNITY

We hold live and virtual events for people with FA and their families to learn, connect, meet with experts, and participate in research opportunities.