UT Southwestern Harold C. Simmons Comprehensive Cancer Center | NCI Designated Comprehensive Cancer Center

Fanconi Anemia (FA)

What you should know about Fanconi Anemia

Fanconi Anemia (FA) is a genetic condition caused by mutations in at least one of 16 genes that are involved in DNA repair. Because individuals with FA do not have the same DNA repair as the general population, they have increased risks to have bone marrow failure (aplastic anemia), physical abnormalities and cancer. Physical differences in individuals with FA include: being short in stature, having unique skin markings, experiencing abnormal development of their bones, and being more likely to have problems with their eyes, kidneys and urinary tract, ears, heart, gastrointestinal system, and central nervous system (CNS).

Cancer Risks Associated with Fanconi Anemia

People with FA are at an increased risk to develop cancers of the blood, head and neck, skin, gastrointestinal (GI), and gynecologic systems. Blood cancers in FA are generally caused by abnormalities in the blood-forming cells of the bone marrow. The most common types of blood cancers in individuals with FA are acute myeloid leukemia (AML) and myelodysplastic syndrome (MDS). Overall, people with FA have between a 10-30% risk to develop one of these types of cancer, and these cancers tend to occur at younger ages than in the general population.

Risks to Family Members

There are at least 16 genes that have been associated with FA. Because there are multiple genes associated with this condition, the risk for family members is based on the gene (or genes) in which a family has a mutation(s). Different genes are associated with different types of inheritance and at least three common types of inheritance patterns have been observed in families with FA: autosomal dominant (AD), autosomal recessive (AR) and X-linked inheritance. We recommend speaking with a geneticist or genetic counselor to discuss the risks for family members and inheritance patterns.

Resources & Managing Cancer Risks

The Fanconi Anemia Research Fund publishes guidelines for diagnosis and management of Fanconi Anemia which are freely available via their website at https://www.fanconi.org/. Individuals with FA should be under the care of a specialist familiar with managing the condition.

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