Genetics Uncoded:

NxGen MDx

Facts about *FANCC*-Associated Fanconi Anemia

What Your Test Results Mean

Carriers show no symptoms of FANCC-associated Fanconi anemia (FANCC-associated FA) and are not at risk to develop symptoms of the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

FANCC-Associated Fanconi Anemia Explained

FANCC-associated FA is an inherited disorder characterized by a broad range of physical abnormalities, progressive bone marrow failure, and increased risk of cancer. Physical abnormalities, found in 75% of cases, are typically present at birth and commonly include short stature, upper limb abnormalities, and hyperpigmentation of the skin. Hematologic complications of FA typically occur within the first decade of life but are highly variable. Cancer risk remains a concern throughout the lifespan. Individuals with FA are not able to properly repair DNA damage due to defects in the *FANCC* gene leading to premature cell death or uncontrolled cell growth.

Monitoring for evidence of bone marrow failure and solid tumors as well as growth and development is recommended surveillance for individuals with FA. Treatment is generally symptomatic; however, some individuals undergo hematopoietic stem cell transplantation to cure the hematologic manifestations of FA. Stem cell transplant does not reduce and may increase the risk of solid tumors. The majority of individuals with FA reach adulthood. Lifespan is variable with median age of death in the thirties.

How the Genetics Work

There are at least 16 genes associated with Fanconi anemia. This testing identifies pathogenic variants in one of these genes, *FANCC*. Variants in this gene cause approximately 14% of Fanconi anemia. *FANCC*-associated FA is caused by variants in the *FANCC* gene. In general, individuals have two copies of the *FANCC* gene. Carriers have a single variant in the *FANCC* gene while individuals with *FANCC*-associated FA have variants in both copies of *FANCC*, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

NxGenMDx, LLC
801 Broadway Ave NW, Suite 203
Grand Rapids, MI, 49504
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