Fanconi Anemia Type C

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Fanconi anemia type C. There is preliminary evidence suggesting that carriers of Fanconi anemia type C may have a higher chance of developing cancer, but the evidence is currently limited. You and your partner or donor would both have to be carriers of Fanconi anemia type C for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

We recommend that you share this information with all of your health care providers. Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended. If both you and your partner or donor are carriers for Fanconi anemia type C, each of your children has a 1 in 4 (25%) chance to have the condition. Consultation with a genetic counselor for a more detailed risk assessment is available.

Fanconi Anemia Type C Explained

What is Fanconi Anemia Type C?

Fanconi anemia type C is an inherited disorder characterized by a range of physical abnormalities, progressive bone marrow failure, and an increased risk of cancer. Physical abnormalities, found in 75% of cases, are typically present at birth and include short stature, upper limb abnormalities, and hyperpigmentation of the skin. The hematologic complications of Fanconi anemia type C typically develop within the first decade of life and are highly variable. Cancer risk remains a concern throughout the lifespan. Individuals with Fanconi anemia type C are not able to properly repair DNA damage, leading to premature cell death or uncontrolled cell growth.

Prognosis

Prognosis is generally unfavorable. Due to the physical abnormalities, bone marrow failure, and increased risk for cancer, affected individuals typically die before the age of 30.

Treatment

Hematopoietic stem cell transplantation (HSCT) is the only curative therapy for the hematologic manifestations of Fanconi anemia, but the high risk for solid tumors remains and may even be increased in those undergoing HSCT. Surveillance for early detection and surgical removal remain the mainstay of treatment for solid tumors. Oral androgens improve blood cell counts (red cell, white cell, and platelets) in many individuals.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/fanconi-anemia

National Society of Genetic Counselors
https://www.nsgc.org/

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