NxGen MDx

Congenital Amegakaryocytic Thrombocytopenia

What Your Results Mean

Test results indicate that you are a carrier of congenital amegakaryocytic thrombocytopenia (CAMT). Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of CAMT 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.

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 in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for congenital amegakaryocytic thrombocytopenia, each of your children has a 1 in 4 (25%) chance to have the condition.

Congenital Amegakaryocytic Thrombocytopenia Explained

What is Congenital Amegakaryocytic Thrombocytopenia?

Congenital amegakaryocytic thrombocytopenia (CAMT) is an inherited condition that causes bone marrow failure. Symptoms of CAMT first begin to show during infancy, often during the first month of life. CAMT is characterized by excessive, life-threatening bleeding, bruising, and tiny red dots under the skin that result from small bleeds into the skin known as petechiae. Affected individuals may also experience central nervous abnormalities, cardiac defects, and psychomotor delays.

Prognosis

With supportive treatment, bone marrow failure occurs around the first decade of life. Hematopoietic stem cell transplant (HSCT) is considered curative and is successful for over 50% of affected individuals. Thirty percent of patients with CAMT die from bleeding complications before the HSCT and 20% die due to the HSCT.

Treatment

Treatment for CAMT may include bone marrow or hematopoietic stem cell transplant. Affected individuals may also benefit from blood transfusions.

Resources

Genetic and Rare Diseases Information Center (GARD)
https://rarediseases.info.nih.gov/diseases/640/congenital-amegakaryocytic-thrombocytopenia

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

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