Genetics Uncoded:

NxGen MDx

Facts about

Congenital Amegakaryocytic Thrombocytopenia

What Your Test Results Mean

Carriers show no symptoms of congenital amegakaryocytic thrombocytopenia (CAMT) and are not at risk to develop CAMT. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Congenital Amegakaryocytic Thrombocytopenia Explained

CAMT is an inherited condition that causes bone marrow failure. The *MPL* gene provides instructions for making the thrombopoietin receptor protein, which promotes the growth and division of cells. This receptor is especially important for the proliferation of certain blood cells called megakaryocytes, which produce platelets. Lack of thrombopoietin receptor protein causes excessive bleeding and bruising. Most children with CAMT exhibit normal neurologic development and normal physical and developmental growth, but may have decreased lifespan due to their health complications. Treatment for CAMT may include bone marrow or hematopoietic stem cell transplants.

How the Genetics Work

CAMT is an autosomal recessive condition caused by variants in the *MPL* gene. In general, individuals have two copies of the *MPL* gene. Carriers of CAMT have a single variant in one copy of the *MPL* gene while individuals with CAMT have variants in both copies of their genes, 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

Revised 12/15/16