Genetics Uncoded:

NxGen MDx

Facts about Bloom Syndrome

What Your Test Results Mean

Carriers typically show no symptoms of Bloom syndrome; however, carriers are at an increased risk of having a child with Bloom syndrome. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Bloom Syndrome Explained

Bloom syndrome is an inherited disorder characterized by proportionate short stature from the prenatal period to adulthood. Other features are variable from individual to individual and may include learning disabilities, sensitivity to the sun, gastroesophageal reflux, recurrent ear and lung infections, infertility in males, and an increased risk for cancer. Individuals with Bloom syndrome are not able to maintain the stability of genetic material in their bodies due to the defect in the *BLM* gene. The cause of the main features of Bloom syndrome are not known; however, the instability of genetic material is associated with cancer predisposition.

Treatment of individuals with Bloom syndrome is symptomatic. Individuals with Bloom syndrome should avoid sun exposure and undergo increased surveillance for cancer starting in childhood as cancer is the greatest cause of death in individuals with Bloom syndrome. Lifespan is variable but is typically decreased in individuals with a cancer diagnosis.

How the Genetics Work

Bloom syndrome is an autosomal recessive disorder caused by variants in the *BLM* gene. In general, individuals have two copies of the *BLM* gene. Carriers of Bloom syndrome have a single variant in *BLM* while individuals with Bloom syndrome have variants in both copies of *BLM*, 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.

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