What Your Test Results Mean

Test results indicate that you are a carrier of Bloom syndrome. Carriers typically show no symptoms of Bloom syndrome; however, carriers are at an increased risk of having a child with Bloom syndrome. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

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.

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How the Genetics Work

The clinical features of Bloom syndrome are caused by mutations in the BLM gene. Individuals have two copies of the BLM gene. Carriers of Bloom syndrome have a single mutation in BLM while individuals with Bloom syndrome have mutations in both copies of BLM, one inherited from each parent.

Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

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.