



Facts about Nijmegen Breakage Syndrome



What Your Test Results Mean

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

● Nijmegen Breakage Syndrome Explained

Nijmegen breakage syndrome is an inherited condition characterized by short stature, an unusually small head size, distinctive facial features, an increased risk of cancer, and immunodeficiency. The symptoms of the disorder are caused by a non-functional protein, nibrin. Nibrin is normally responsible for repairing damaged DNA in a person's cells, and when it is non-functional, the cells accumulate genetic damage. Individuals with Nijmegen breakage syndrome are very susceptible to recurrent infections, especially of the respiratory tract. These individuals also develop mild to moderate intellectual disability, and most affected women have primary ovarian insufficiency. Most individuals with Nijmegen breakage syndrome develop some type or multiple types of cancer in childhood or adolescence. Due to the increased risk for cancer and recurrent infections, lifespan is limited. Treatment is symptomatic.

● How the Genetics Work

The clinical features of Nijmegen breakage syndrome can be explained by variants in the *NBN* gene. In general, individuals have two copies of the *NBN* gene. Carriers of Nijmegen breakage syndrome have a single variant in one copy of the *NBN* gene while individuals with Nijmegen breakage syndrome 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 **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.