NxGen MDx

Nijmegen Breakage Syndrome

What Your Results Mean

Test results indicate that you are a carrier of Nijmegen breakage syndrome. Carriers typically show no symptoms of disease; however, recent studies have shown that some carriers may be at a greater than average risk of developing breast and ovarian cancer. Risk for 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.

We recommend that you share this information with all of your healthcare providers. Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

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 Nijmegen breakage syndrome, each of your children has a 1 in 4 (25%) chance to have the condition.

Nijmegen Breakage Syndrome Explained

What is Nijmegen Breakage Syndrome?

Nijmegen breakage syndrome (NBS) is an inherited condition characterized by short stature, an unusually small head size, distinctive facial features, an increased risk of cancer, and immunodeficiency. In this disease, the NBN gene responsible for repairing broken DNA is defective. As a result, affected individuals are sensitive to the effects of radiation exposure and other agents that can cause breaks in DNA. Intellect appears to develop normally or near-normally in early childhood, but typically declines until the person reaches mild-to-moderate levels of mental disability around the age of 10. People with NBS can live into adulthood, but typically not beyond their thirties or forties.

Prognosis

Prognosis is generally unfavorable. Affected individuals have a malfunctioning immune system and are susceptible to recurrent infections such as bronchitis, pneumonia, and sinusitis. Additionally, affected individuals are 50 times more likely to develop cancer than people without the condition. About half of patients develop non-Hodgkin's lymphoma by the age of 15 years. Premature death usually occurs from complications of infection and/or malignancy.

Treatment

Treatments are symptomatic, not curative. Due to the specific basic defect underlying immunodeficiency and sensitivity to radiation, patients with NBS require multidisciplinary medical management and long-term follow-up. Intravenous Ig is used to treat immunodeficiency. Vitamin E and folic acid supplementation is also recommended.

Resources

American Cancer Society
https://www.cancer.org/
National Cancer Institute
https://www.cancer.gov/
National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

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