ADA-Related Conditions

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of *ADA*-related conditions. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of *ADA*-related conditions for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

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 are carriers for *ADA*-related conditions each of your children has a 1 in 4 (25%) chance to have the condition.

ADA-Related Conditions Explained

What are *ADA*-Related Conditions?

ADA-related conditions or adenosine deaminase (ADA) deficiency describes a group of metabolic conditions characterized by damage to the immune system. It is also a common cause of severe combined immunodeficiency (SCID). Reduced or eliminated activity of adenosine deaminase allows the buildup of deoxyadenosine to reach levels that are toxic to lymphocytes. Loss of these lymphocytes results in the signs and symptoms of SCID. As such, there are three different forms of adenosine deaminase (ADA) deficiency.

Adenosine Deaminase-Deficient Severe Combined Immunodeficiency Disease (SCID)

ADA-SCID is the most severe form of *ADA*-related conditions. The diagnosis of this form is often made within the first six months of life. Signs and symptoms can include recurrent pneumonia, severe diarrhea, and inflammation of the skin. Individuals may additionally experience growth failure, as well as neurological and skeletal abnormalities.

Delayed/Late-Onset Adenosine Deaminase Deficiency

Delayed or late-onset ADA Deficiency is typically diagnosed between the ages of one and 10 years old. Rarely, a diagnosis may be made in the second or third decade of life. Symptoms are less severe than with ADA-SCID. Other symptoms can include but are not limited to common upper respiratory tract infections, ear infections, plantar and palmar warts, and allergies.

Partial Adenosine Deaminase Deficiency

Partial ADA is considered to be a benign condition that is consistent with normal immune response. Individuals with partial ADA typically only present with low levels of ADA enzymes in the blood.

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

Revised: 4/2022

Prognosis

Prognosis depends on the severity of an *ADA*-related condition diagnosis, how early treatment is started, and the response to treatment. With treatment and a successful hematopoietic stem cell transplant, individuals with ADA-SCID have about a 90% chance of survival for at least one year if the transplant is successful. Overall, the long-term outlook is considered to be generally good; however, without early diagnosis and treatment, babies with ADA deficiency usually do not survive past the age of two.

Treatment

The primary form of treatment is a hematopoietic stem cell transplant (HSCT). If a transplant is not possible, enzyme replacement therapy can be considered. Gene therapy is still considered to be experimental in the United States.

Resources

National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/severe-combined-immunodeficiency/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/adenosine-deaminase-deficiency
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

Revised: 4/2022