Ataxia-Telangiectasia

What Your Results Mean

Testing results indicate that you are a carrier of ataxia-telangiectasia. Carriers of ataxia-telangiectasia are not expected to show signs and symptoms of the disease; however, they are at an increased risk to develop cancer, specifically breast cancer. It is recommended that carriers of ataxia-telangiectasia speak to a genetic counselor that specializes in cancer genetics regarding these risks. 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 and discuss this information with all of your health care 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. If both you and your partner are both carriers for ataxia-telangiectasia, each of your children has a 1 in 4 (25%) chance to have the condition. Consultation with a genetic counselor for a more detailed risk assessment is available.

Ataxia-Telangiectasia Explained

What is Ataxia-Telangiectasia?

Ataxia-telangiectasia is an inherited neurodegenerative condition that primarily affects the nervous system and immune system. Symptoms include difficulty coordinating movements (ataxia), spidery red skin lesions caused by dilated blood vessels, a weakened immune system that may lead to recurrent infections, and an increased risk for cancer. Individuals with ataxia-telangiectasia may additionally have slurred speech and have trouble moving their eyes from one side to the other. They are also extremely sensitive to radiation, especially medical x-rays.

Prognosis

Affected individuals typically have a decreased lifespan but most live into their second decade of life. Although rare, some individuals live until their fourth decade of life. Most individuals are wheelchair-bound by the age of 10.

Treatment

Treatment for ataxia-telangiectasia is symptomatic; there is currently no cure. Individuals may benefit from injections to boost the immune system. Physical, occupational, and speech therapy can additionally be considered.

Resources

A-T Children's Project
https://www.atcp.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/ataxia-telangiectasia
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/ataxia-telangiectasia/
National Society of Genetic Counselors
https://www.nsgc.org/

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