Ataxia Telangiectasia

Ataxia telangiectasia is a rare neurodegenerative disease caused by a defect in the ATM gene, which is responsible for recognizing double-strand breaks in DNA and recruiting the appropriate proteins to fix the break. The hallmarks of this disease are ataxia, which refers to poor coordination due to damage of the cerebellum and telangiectasia, which are small dilated blood vessels also commonly called spider angiomas. Inability to repair defects in DNA makes individuals extremely sensitive to radiation, which can damage DNA. This gene is a tumor suppressor gene and loss of function leads to increased risk of cancer, especially lymphomas and leukemias. Individuals also commonly have IgA deficiencies, making them susceptible to infections of the ears, sinuses, and lungs.



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Pathophysiology

Autosomal Recessive
Recessive-chocolate
Ataxia telangiectasia is inherited in an autosomal recessive modality. This means two copies of the abnormal gene must be present in order for the disease to develop.

ATM Gene
ATM
The classical form of A-T results from the presence of two A-T mutated (ATM) genes on chromosome 11 leading to total loss of the ATM protein (a protein kinase).

Defect in DNA repair
Defective DNA Repair-man falling
This disease is caused by a defect in the ATM gene, which is responsible for recognizing double-strand breaks in DNA and recruiting the appropriate proteins to fix the break.

Signs & Symptoms

Cerebellar defects
Silver-cerebellum-bell
One of the hallmarks of this disease include cerebellar defects, which can lead to ataxia.

Spider Angiomas
Spider Angel
Spider angiomas are a type of telangiectasia found slightly beneath the surface of the skin. These contain a central red spot and reddish extensions that radiate outwards like a spider's web.

Sensitive to Radiation
Radio-waves damaging DNA
Inability to repair defects in DNA makes individuals extremely sensitive to radiation, which can cause double-stranded breaks in DNA. Individuals with ataxia telangiectasia should not undergo procedures involving radiation unless absolutely critical.

Defective Tumor Suppressor Gene
Defective Tumor-suppressing-cage releasing Tumor-guy
This gene is a tumor suppressor gene and loss of function leads to increased risk of cancer, especially lymphomas and leukemias.

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Increased risk of Cancer, Leukemias, and Lymphomas

Tumor-guy, Leukemia-Luke, and Lime-foam

People with Ataxia telangiectasia have increased incidence of cancers, especially lymphomas and leukemia due to loss of the tumor suppressor gene ATM. Treatment of their cancers should avoid use of radiation therapy and chemotherapy drugs that work similarly to radiation therapy as these are extremely toxic for people with this disease.

Increased AFP levels

Up-arrow Association of Frying Pans

Patients with ataxia telangiectasia present with increased levels of AFP. Most patients have increased levels after the age of 2, and these levels are especially high in the newborn.

IgA deficiency

(IgA) Apple-goblin deficiency (trampled)

About two thirds of people with this disease have abnormalities of the immune system with an IgA deficiency being the most common. This makes individuals susceptible to repeated sino pulmonary infections.

Repeated sinopulmonary infections

Sinner with big nose

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