Ataxia Telangiectasia

Ataxia telangiectasia (A-T) is a rare, progressive, childhood disease that affects the nervous system and other body systems.

SYMPTOMS

The first signs of the disease usually occur during the first decade of life. First symptoms include lack of balance and slurred speech. The hallmarks of A-T are ataxia (lack of muscle control) and telangiectasias (tiny, red "spider" veins). These "spider" veins appear in the corners of the eyes or on the surface of the ears and cheeks soon after the onset of ataxia.

Individuals with A-T are predisposed to leukemia and lymphoma. Individuals with A-T are extremely sensitive to radiation exposure. Many individuals with A-T have a weakened immune system. They are susceptible to recurrent respiratory infections. Other features of the disease may include:

- Mild diabetes mellitus.
- Early graying of the hair.
- Difficulty swallowing, which causes choking and/or drooling.
- Slowed growth.

Children with A-T usually maintain normal or above normal intelligence.

TREATMENT

There is no cure for A-T. There is no way to slow the progression of the disease. Treatment is symptomatic and supportive. Physical and occupational therapy may help maintain flexibility. Speech therapy may be needed. Gamma-globulin injections may be given to help supplement the weakened immune systems. High-dose vitamin regimes may also be used.

WHAT WILL HAPPEN

The *prognosis* (likely outcome) for individuals with A-T is poor. Those with the disease usually die in their teens or early 20s.

RESEARCH BEING DONE

NINDS-supported researchers recently discovered the A-T gene. The discovery of the gene may lead to more accurate *diagnosis* (learning what is wrong) of the disease and the development of effective treatments.

Research is continually being done to learn more about this problem.

Provided by: The National Institute of Neurological Disorders and Stroke

Document Released: 12/08/2003 Document Re-Released: 06/30/2008

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