TTPA Gene

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Alpha tocopherol transfer protein.

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1. Normal Function

The *TTPA* gene provides instructions for making the α-tocopherol transfer protein (αTTP), which is found in the liver and brain. This protein controls the distribution of vitamin E obtained from the diet (also called α-tocopherol) to cells and tissues throughout the body. Vitamin E is an antioxidant that protects cells in the body from the damaging effects of unstable molecules called free radicals. Normally, vitamin E derived from food is absorbed in the intestine and then transported into the liver on molecules called chylomicrons. After a meal, chylomicrons are formed to transport fat-soluble vitamins (such as vitamin E), dietary fats, and cholesterol from the intestine to the liver. Once in the liver, αTTP transfers vitamin E from chylomicrons to very low-density lipoproteins (VLDLs), which carry fat, fat-soluble vitamins, and cholesterol from the liver to other tissues throughout the body. The VLDLs are then released into the bloodstream so the accompanying vitamin E can be used in the body. The αTTP protein is also thought to transport vitamin E to nerve cells (neurons) in the brain.

2. Health Conditions Related to Genetic Changes

2.1. Ataxia with Vitamin E Deficiency

More than 20 mutations in the *TTPA* gene have been found to cause ataxia with vitamin E deficiency. This condition is characterized by the development of neurological problems including difficulty coordinating movements (ataxia) due to a buildup of harmful molecules called free radicals. Some of these mutations cause no functional protein to be made, while others change a single protein building block (amino acid) in the αTTP protein, reducing its function. As a result, the body cannot retain or use dietary vitamin E, which leads to reduced levels of this vitamin in the blood and the accumulation of free radicals. One *TTPA* gene mutation that is found in the Japanese population changes the amino acid histidine to the amino acid glutamine at position 101 in the αTTP protein (written as His101Glu or H101Q). This mutation is associated with the development of an eye disorder called retinitis pigmentosa that causes vision loss in people with ataxia with vitamin E deficiency.

Mutations in the *TTPA* gene that cause no functional αTTP protein to be made are associated with a severe form of ataxia that begins at a young age. Mutations that reduce but do not eliminate the protein's function are associated with milder ataxia that occurs at a later age and progresses more slowly.

3. Other Names for This Gene

- alpha-tocopherol transfer protein
- alphaTTP
- ataxia (Friedreich-like) with vitamin E deficiency
- ATTP
- AVED
- tocopherol (alpha) transfer protein
- TTP1

- TTPA_HUMAN

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