ELP1 Gene

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Elongator complex protein 1: The ELP1 gene provides instructions for making a protein called elongator complex protein 1 (ELP1).

Keywords: genes

1. Normal Function

This protein is found in a variety of cells throughout the body, including brain cells. It is part of a six-protein complex called the elongator complex. The elongator complex plays a key role in transcription, the process that transfers information in genes to the cell machinery that makes proteins. Researchers believe that the elongator complex is important for the transcription of proteins that affect the cell's structural framework (the cytoskeleton) and cell movement (motility). The cytoskeleton and cell motility are essential for the growth and development of cells. For example, the cytoskeleton plays a critical role in the growth of nerve cells, particularly the specialized extensions called axons and dendrites that are required for the transmission of nerve impulses. Cell motility is crucial for the movement of nerve cells to their proper locations in the brain.

2. Health Conditions Related to Genetic Changes

2.1 Familial Dysautonomia

Nearly all individuals with familial dysautonomia have two copies of the same mutation in each cell. This mutation can disrupt how information in the *ELP1* gene is spliced together during transcription. As a result of this splicing error, a reduced amount of ELP1 protein is produced. This mutation behaves inconsistently, however. Some cells produce near normal amounts of ELP1 protein, and other cells—particularly brain cells—have very little of the protein.

In a small number of reported familial dysautonomia cases, researchers have identified other mutations that change one of the protein building blocks (amino acids) in the ELP1 protein. In these cases, the amino acid arginine is replaced by the amino acid proline at position 696 (written as Arg696Pro), or the amino acid proline is replaced by the amino acid leucine at position 914 (written as Pro914Leu). People with one of these improper amino acid substitutions also have the splicing mutation. Together, these mutations impair the ELP1 protein.

It is unclear how *ELP1* gene mutations lead to the signs and symptoms of familial dysautonomia. Reduced amounts of ELP1 protein may impair the growth and development of nerve cells by disrupting the cytoskeleton and cell motility.

3. Other Names for This Gene

- DYS
- ELP1_HUMAN
- IKAP
- IKBKAP
- IKI3
- inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein
- TOT1

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