

ELP1 gene

elongator acetyltransferase complex subunit 1

Normal Function

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

Health Conditions Related to Genetic Changes

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.

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

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ELP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8518\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8518[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28IKBKAP%5BTIAB%5D%29+OR+%28%28IKAP%5BTIAB%5D%29+OR+%28ELP1%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- ELONGATOR COMPLEX PROTEIN 1; ELP1 (<https://omim.org/entry/603722>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8518>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ELP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ELP1[gene]))

References

- Anderson SL, Coli R, Daly IW, Kichula EA, Rork MJ, Volpi SA, Ekstein J, Rubin BY. Familial dysautonomia is caused by mutations of the IKAP gene. Am J Hum Genet. 2001 Mar;68(3):753-8. doi: 10.1086/318808. Epub 2001 Jan 22. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11179021>) or Free article on PubMed Central (<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC11179021/>)

s://www.ncbi.nlm.nih.gov/pmc/articles/PMC1274486/)

- Axelrod FB. Familial dysautonomia. Muscle Nerve. 2004 Mar;29(3):352-63. doi:10.1002/mus.10499. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14981733>)
- Close P, Hawkes N, Cornez I, Creppe C, Lambert CA, Rogister B, Siebenlist U, Merville MP, Slaugenhaupt SA, Bours V, Svejstrup JQ, Chariot A. Transcription impairment and cell migration defects in elongator-depleted cells: implication for familial dysautonomia. Mol Cell. 2006 May 19;22(4):521-31. doi:10.1016/j.molcel.2006.04.017. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16713582>)
- Cuajungco MP, Leyne M, Mull J, Gill SP, Lu W, Zagzag D, Axelrod FB, Maayan C, Gusella JF, Slaugenhaupt SA. Tissue-specific reduction in splicing efficiency of IKBKAP due to the major mutation associated with familial dysautonomia. Am J Hum Genet. 2003 Mar;72(3):749-58. doi: 10.1086/368263. Epub 2003 Feb 6. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12577200>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1180251/>)
- Ibrahim EC, Hims MM, Shomron N, Burge CB, Slaugenhaupt SA, Reed R. Weak definition of IKBKAP exon 20 leads to aberrant splicing in familial dysautonomia. Hum Mutat. 2007 Jan;28(1):41-53. doi: 10.1002/humu.20401. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16964593>)
- Leyne M, Mull J, Gill SP, Cuajungco MP, Oddoux C, Blumenfeld A, Maayan C, Gusella JF, Axelrod FB, Slaugenhaupt SA. Identification of the first non-Jewish mutation in familial Dysautonomia. Am J Med Genet A. 2003 May 1;118A(4):305-8. doi: 10.1002/ajmg.a.20052. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12687659>)
- Slaugenhaupt SA, Blumenfeld A, Gill SP, Leyne M, Mull J, Cuajungco MP, Liebert CB, Chadwick B, Idelson M, Reznik L, Robbins C, Makalowska I, Brownstein M, Krappmann D, Scheidereit C, Maayan C, Axelrod FB, Gusella JF. Tissue-specific expression of a splicing mutation in the IKBKAP gene causes familial dysautonomia. Am J Hum Genet. 2001 Mar;68(3):598-605. doi: 10.1086/318810. Epub 2001 Jan 22. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/1179008>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1274473/>)
- Slaugenhaupt SA, Gusella JF. Familial dysautonomia. Curr Opin Genet Dev. 2002 Jun;12(3):307-11. doi: 10.1016/s0959-437x(02)00303-9. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12076674>)

Genomic Location

The *ELP1* gene is found on chromosome 9 (<https://medlineplus.gov/genetics/chromosome/9/>).

Last updated May 1, 2007