FIP1L1 gene

FIP1 like 1 (S. cerevisiae)

Normal Function

The *FIP1L1* gene provides instructions for making part of a protein complex named cleavage and polyadenylation specificity factor (CPSF). This complex of proteins plays an important role in processing molecules called messenger RNAs (mRNAs), which serve as the genetic blueprints for making proteins. The CPSF protein complex helps add a string of the RNA building block adenine to the mRNA, creating a polyadenine tail or poly(A) tail. The poly(A) tail is important for stability of the mRNA and for protein production from the blueprint.

Health Conditions Related to Genetic Changes

PDGFRA-associated chronic eosinophilic leukemia

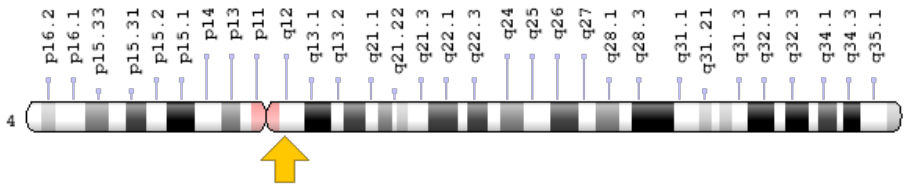
A deletion of genetic material from chromosome 4 brings together part of the *FIP1L1* gene and part of another gene called *PDGFRA*, creating the *FIP1L1-PDGFRA* fusion gene. This mutation is a somatic mutation, which means it is acquired during a person's lifetime and is present only in certain cells. This fusion gene causes *PDGFRA*-associated chronic eosinophilic leukemia, which is a type of blood cell cancer characterized by an increased number of eosinophils, a type of white blood cell involved in allergic reactions.

The FIP1L1-PDGFRA protein produced from the fusion gene has the function of the normal PDGFRA protein, which stimulates signaling pathways inside the cell that control many important cellular processes, such as cell growth and division (proliferation) and cell survival. Unlike the normal PDGFRA protein, however, the FIP1L1-PDGFRA protein is constantly turned on (constitutively activated), which means the cells are always receiving signals to proliferate. When the *FIP1L1-PDGFRA* fusion gene occurs in blood cell precursors, the growth of eosinophils (and occasionally other blood cells) is poorly controlled, leading to *PDGFRA*-associated chronic eosinophilic leukemia. It is unclear why eosinophils are preferentially affected by this genetic change.

Chromosomal Location

Cytogenetic Location: 4q12, which is the long (q) arm of chromosome 4 at position 12

Molecular Location: base pairs 53,377,572 to 53,462,611 on chromosome 4 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FIP1-like 1 protein
- FIP1_HUMAN
- hFip1
- pre-mRNA 3'-end-processing factor FIP1
- Rhe

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): Processing of mRNA in Eukaryotes
  https://www.ncbi.nlm.nih.gov/books/NBK9864/#A1031

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28FIP1L1%5BTIAB%5D%29+OR+%28FIP1+like+1%5BTIAB%5D%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

Catalog of Genes and Diseases from OMIM

- FIP1-LIKE 1
  http://omim.org/entry/607686

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/FIP1L1ID40577ch4q12.html
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:19124
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:81608
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/81608
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