FLI1 gene
Fli-1 proto-oncogene, ETS transcription factor

Normal Function

The FLI1 gene provides instructions for making the FLI protein, which controls the activity (transcription) of genes. Transcription is the first step in the process of producing proteins. The FLI protein is part of a group of related proteins, called the Ets family of transcription factors, that control transcription. The FLI protein attaches (binds) to certain regions of DNA and turns on (activates) the transcription of nearby genes. The proteins produced from these genes control many important cellular processes, such as cell growth and division (proliferation), maturation (differentiation), and survival. The FLI protein is found primarily in blood cells and is thought to regulate their development.

Health Conditions Related to Genetic Changes

Ewing sarcoma

Mutations involving the FLI1 gene cause a type of cancerous tumor known as Ewing sarcoma. These tumors develop in bones or soft tissues such as nerves and cartilage. There are several types of Ewing sarcoma, including Ewing sarcoma of bone, extraosseous Ewing sarcoma, peripheral primitive neuroectodermal tumor, and Askin tumor. The mutations that cause these tumors are acquired during a person's lifetime and are present only in the tumor cells. This type of genetic change, called a somatic mutation, is not inherited. The most common mutation that causes Ewing sarcoma is a rearrangement (translocation) of genetic material between chromosome 11 and chromosome 22. This translocation, written as t(11;22), fuses part of the FLI1 gene on chromosome 11 with part of another gene called EWSR1 on chromosome 22, creating an EWSR1/FLI1 fusion gene.

The protein produced from the EWSR1/FLI1 fusion gene, called EWS/FLI, has functions of the protein products of both genes. Like the FLI protein, the EWS protein, produced from the EWSR1 gene, can regulate transcription. The EWS/FLI protein has the DNA-binding function of the FLI protein as well as the transcription regulation function of the EWS protein. It is thought that the EWS/FLI protein turns the transcription of a variety of genes on and off abnormally. This dysregulation of transcription leads to uncontrolled growth and division (proliferation) and abnormal maturation and survival of cells, causing tumor development.

The EWSR1/FLI1 fusion gene occurs in approximately 85 percent of Ewing sarcomas. The remaining tumors result from translocations that fuse the EWSR1 gene with other genes.
Jacobsen syndrome

Chromosomal Location
Cytogenetic Location: 11q24.3, which is the long (q) arm of chromosome 11 at position 24.3
Molecular Location: base pairs 128,685,263 to 128,813,267 on chromosome 11 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Other Names for This Gene
- EWSR2
- FLI1_HUMAN
- Friend leukemia integration 1 transcription factor
- Friend leukemia virus integration 1
- proto-oncogene Fli-1
- SIC-1
- transcription factor ERGB

Additional Information & Resources
Scientific Articles on PubMed
- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28FLI1%5BTIAB%5D%29+OR+%28Friend+leukemia+vir+integration+1%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM
- FRIEND LEUKEMIA VIRUS INTEGRATION 1
  http://omim.org/entry/193067
Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/FLI1ID79ch11q24.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FLI1%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2313
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q01543

Sources for This Summary

- OMIM: FRIEND LEUKEMIA VIRUS INTEGRATION 1
  http://omim.org/entry/193067
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/7517940
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  https://ghr.nlm.nih.gov/gene/FLI1

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