SETBP1 gene
SET binding protein 1

Normal Function

The \textit{SETBP1} gene provides instructions for making a protein called SET binding protein 1 (SETBP1), which is found in cells throughout the body. The SETBP1 protein is known to attach (bind) to another protein called SET. However, the function of the SETBP1 protein and the effect of its binding to the SET protein are unknown.

Health Conditions Related to Genetic Changes

\textbf{Schinzel-Giedion syndrome}

At least five mutations in the \textit{SETBP1} gene have been identified in children with Schinzel-Giedion syndrome, a severe condition apparent at birth that is characterized by distinctive facial features and abnormalities in many body systems. The gene mutations in affected individuals cluster in one region of the gene known as exon 4. However, the effects of the mutations on the function of the gene or the protein are unknown. Researchers are working to understand how mutations in the \textit{SETBP1} gene cause the signs and symptoms of Schinzel-Giedion syndrome.

Chromosomal Location

\textbf{Cytogenetic Location:} 18q12.3, which is the long (q) arm of chromosome 18 at position 12.3

\textbf{Molecular Location:} base pairs 44,680,072 to 45,068,510 on chromosome 18 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Other Names for This Gene

- KIAA0437
- SEB
- SET-binding protein
- SET-binding protein isoform a
- SET-binding protein isoform b
- SETBP_HUMAN

Additional Information & Resources

Scientific Articles on PubMed
- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SETBP1%5BTIAB%5D%29+OR+%28SET+binding+protein+1%5BTIAB%5D%29%29+OR+%28%28SEB%5BTIAB%5D%29%29+OR+%28SET-binding+protein%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+1080+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM
- SET-BINDING PROTEIN 1
  http://omim.org/entry/611060

Research Resources
- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/SETBP1ID44031ch18q12.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SETBP1%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:26040
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q9Y6X0
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20436468

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11231286

- OMIM: SET-BINDING PROTEIN 1
  http://omim.org/entry/611060

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21371013

Reprinted from Genetics Home Reference:
  https://ghr.nlm.nih.gov/gene/SETBP1

Reviewed: January 2012
Published: November 27, 2018

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