EHMT1 gene

euchromatic histone lysine methyltransferase 1

Normal Function

The *EHMT1* gene provides instructions for making an enzyme called euchromatic histone methyltransferase 1. Histone methyltransferases are enzymes that modify proteins called histones. Histones are structural proteins that attach (bind) to DNA and give chromosomes their shape. By adding a molecule called a methyl group to histones, histone methyltransferases can turn off (suppress) the activity of certain genes, which is essential for normal development and function.

Health Conditions Related to Genetic Changes

Kleefstra syndrome

Kleefstra syndrome, a disorder affecting many parts of the body, is caused by the loss of the *EHMT1* gene or by mutations that disable its function.

Most people with Kleefstra syndrome are missing a sequence of about 1 million DNA building blocks (base pairs) on one copy of chromosome 9 in each cell. The deletion occurs near the end of the long (q) arm of the chromosome at a location designated q34.3, a region containing the *EHMT1* gene. Some affected individuals have shorter or longer deletions in the same region.

The loss of the *EHMT1* gene from one copy of chromosome 9 in each cell is believed to be responsible for the characteristic features of Kleefstra syndrome in people with the 9q34.3 deletion. However, the loss of other genes in the same region may lead to additional health problems in some affected individuals.

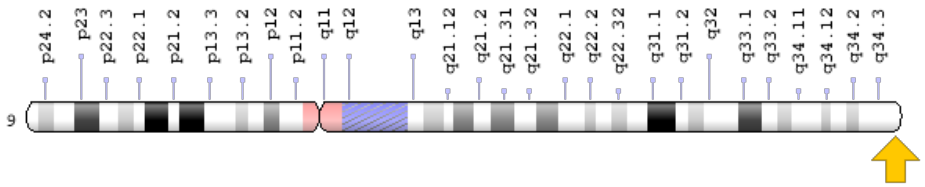
About 25 percent of individuals with Kleefstra syndrome do not have a deletion of genetic material from chromosome 9; instead, these individuals have mutations in the *EHMT1* gene. Some of these mutations change single protein building blocks (amino acids) in euchromatic histone methyltransferase 1. Others create a premature stop signal in the instructions for making the enzyme or alter the way the gene's instructions are pieced together to produce the enzyme. These changes generally result in an enzyme that is unstable and decays rapidly, or that is disabled and cannot function properly.

Either a deletion or a mutation affecting the *EHMT1* gene results in a lack of functional euchromatic histone methyltransferase 1 enzyme. A lack of this enzyme impairs proper control of the activity of certain genes in many of the body's organs and tissues, resulting in the abnormalities of development and function characteristic of Kleefstra syndrome.

Chromosomal Location

Cytogenetic Location: 9q34.3, which is the long (q) arm of chromosome 9 at position 34.3

Molecular Location: base pairs 137,618,992 to 137,836,127 on chromosome 9 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- bA188C12.1
- DEL9q34
- DKFZp667M072
- EHMT1_HUMAN
- Eu-HMTase1
- euchromatic histone-lysine N-methyltransferase 1
- EUHMTASE1
- FLJ12879
- FP13812
- G9a like protein
- G9a-like protein 1
- GLP
- GLP1
- H3-K9-HMTase 5
- histone H3-K9 methyltransferase 5
- histone-lysine N-methyltransferase, H3 lysine-9 specific 5
- KIAA1876
- KMT1D

- lysine N-methyltransferase 1D
- RP11-188C12.1

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Eukaryotic Nuclear DNA Associates with Histone Proteins to Form Chromatin
  https://www.ncbi.nlm.nih.gov/books/NBK21500/#A2248

Clinical Information from GeneReviews

- Kleefstra Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK47079

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28EHMT1%5BTIAB%5D%29+OR+%28%28Eu-HMTase1%5BTIAB%5D%29+OR+%28G9a+like+protein%5BTIAB%5D%29+OR+%28KMT1D%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+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- EUCHROMATIC HISTONE METHYLTRANSFERASE 1
  http://omim.org/entry/607001

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_EHMT1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EHMT1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:24650
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:79813
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/79813
- UniProt
  https://www.uniprot.org/uniprot/Q9H9B1

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