



## EZH2 gene

enhancer of zeste 2 polycomb repressive complex 2 subunit

### Normal Function

The *EZH2* gene provides instructions for making a type of enzyme called a histone methyltransferase. Histone methyltransferases modify proteins called histones, which are structural proteins that attach (bind) to DNA and give chromosomes their shape. By adding a molecule called a methyl group to histones (methylation), histone methyltransferases can turn off (suppress) the activity of certain genes, an essential process in normal development. Specifically, the EZH2 enzyme forms part of a protein group called the polycomb repressive complex-2. By turning off particular genes, this complex is involved in the process that determines the type of cell an immature cell will ultimately become (cell fate determination).

### Health Conditions Related to Genetic Changes

#### Weaver syndrome

More than 30 *EZH2* gene mutations have been identified in people with Weaver syndrome, which involves tall stature, a variable degree of intellectual disability (usually mild), and characteristic facial features. These features can include a broad forehead; widely spaced eyes (hypertelorism); large, low-set ears; a dimpled chin; and a small lower jaw (micrognathia). Some affected individuals have a large head size (macrocephaly). Most of the *EZH2* gene mutations associated with Weaver syndrome change single protein building blocks (amino acids) in the EZH2 enzyme; others insert or delete small amounts of genetic material from the *EZH2* gene, leading to production of an altered EZH2 enzyme. It is unclear how these *EZH2* gene mutations result in the abnormalities characteristic of Weaver syndrome.

#### Prostate cancer

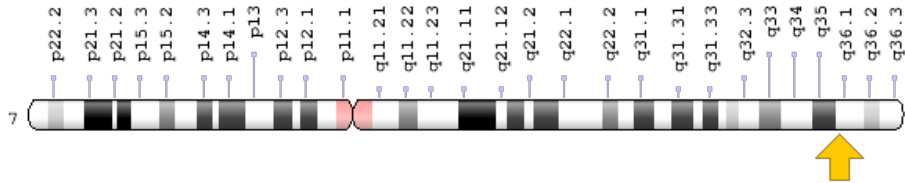
#### Cancers

Changes in the *EZH2* gene have been associated with various types of cancers. Mutations of this gene have been identified in cancers of blood-forming tissues (lymphomas and leukemias). These mutations are described as "gain-of-function" because they appear to enhance the activity of the EZH2 enzyme or give the enzyme a new, atypical function. In addition, excessive activity (overexpression) of the *EZH2* gene has been identified in cancerous tumors of the prostate, breast, and other parts of the body. Changes involving the *EZH2* gene likely impair normal control of cell division (proliferation), allowing cells to grow and divide too fast or in an uncontrolled way and leading to the development of cancer.

## Chromosomal Location

Cytogenetic Location: 7q36.1, which is the long (q) arm of chromosome 7 at position 36.1

Molecular Location: base pairs 148,807,372 to 148,884,349 on chromosome 7 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- enhancer of zeste homolog 2 (Drosophila)
- ENX-1
- EZH2\_HUMAN
- histone-lysine N-methyltransferase EZH2
- KMT6
- KMT6A
- lysine N-methyltransferase 6

## Additional Information & Resources

### Educational Resources

- Madame Curie Bioscience Database: Chromatin Mechanisms Regulating Gene Expression In Health And Disease  
<https://www.ncbi.nlm.nih.gov/books/NBK45032/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EZH2%5BTI%5D%29+OR+%28enhancer+of+zeste+homolog+2%5BTI%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+360+days%22%5Bdp%5D>

## Catalog of Genes and Diseases from OMIM

- ENHANCER OF ZESTE, DROSOPHILA, HOMOLOG 2  
<http://omim.org/entry/601573>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
<http://atlasgeneticsoncology.org/Genes/EZH2ID40517ch7q36.html>
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=EZH2%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:3527](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3527)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:2146>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/2146>
- UniProt  
<https://www.uniprot.org/uniprot/Q15910>

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Reviewed: March 2016  
Published: June 11, 2019

Lister Hill National Center for Biomedical Communications  
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National Institutes of Health  
Department of Health & Human Services