

# EZH2 Gene

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Enhancer of zeste 2 polycomb repressive complex 2 subunit

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## 1. 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).

## 2. Health Conditions Related to Genetic Changes

### 2.1 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.

### 2.2 Prostate cancer

### 2.3 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.

## 3. 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

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