

# NSD2 Gene

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nuclear receptor binding SET domain protein 2

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## 1. Introduction

The *NSD2* gene (also known as *WHSC1* and *MMSET*) provides instructions for making at least three very similar proteins known as MMSET I, MMSET II, and RE-IIBP. These proteins are active both before and after birth in many of the body's cells and tissues. They appear to play an important role in normal development.

At least two of the proteins produced from the *NSD2* gene, MMSET II and RE-IIBP, likely help regulate the activity of other genes. Studies suggest that these proteins function as histone methyltransferases, which are enzymes that modify DNA-associated proteins called histones. By adding a molecule called a methyl group to histones, histone methyltransferases can turn off (suppress) the activity of certain genes. Scientists are working to identify the genes targeted by the MMSET II and RE-IIBP proteins.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Wolf-Hirschhorn syndrome

The *NSD2* gene is located in a region of chromosome 4 that is deleted in people with Wolf-Hirschhorn syndrome. As a result of this deletion, affected individuals are missing one copy of the *NSD2* gene in each cell. A loss of the *NSD2* gene probably disrupts the regulation of several other genes, although these genes have not been identified. Researchers speculate that abnormal gene regulation during development contributes to many of the characteristic features of the disorder, including intellectual disability, growth delay, and a distinctive facial appearance.

### 2.2. Cancers

A chromosomal rearrangement (translocation) involving the *NSD2* gene has been associated with multiple myeloma, a cancer that starts in cells of the bone marrow. This rearrangement is found in 15 to 20 percent of all multiple myelomas. The translocation, which is written as t(4;14)(p16;q32), abnormally fuses the *NSD2* gene on chromosome 4 with part of another gene on chromosome 14. The fusion of these genes overactivates the *NSD2* gene, which appears to promote the uncontrolled growth and division of cancer cells.

## 3. Other Names for This Gene

- FLJ23286
- IL5 promoter REII region-binding protein
- KIAA1090
- MGC176638
- MMSET
- multiple myeloma SET domain protein
- NSD2\_HUMAN
- Nuclear SET domain-containing protein 2
- Probable histone-lysine N-methyltransferase NSD2
- Protein trithorax-5
- REIIBP
- trithorax/ash1-related protein 5

- TRX5
- WHSC1
- Wolf-Hirschhorn syndrome candidate 1

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