# HESX1 Gene

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HESX homeobox 1

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## 1. Introduction

The *HESX1* gene provides instructions for producing a protein that regulates the activity of other genes. On the basis of this action, the HESX1 protein is called a transcription factor. The *HESX1* gene is part of a family of homeobox genes, which act during early embryonic development to control the formation of many body structures.

The HESX1 protein plays an important role in early brain development. In particular, it is essential for the formation of the pituitary, which is a gland at the base of the brain that produces several hormones. The HESX1 protein is also necessary for the development of structures at the front of the brain (the forebrain), including the nerves that carry visual information from the eyes to the brain (optic nerves).

The HESX1 protein interacts with other proteins, including the protein produced from the *PROP1* gene, during embryonic development. Both the HESX1 protein and the PROP1 protein bind to DNA and control the activity of other genes. The HESX1 protein turns off (represses) genes, while the PROP1 protein turns on (activates) genes. These proteins work together to coordinate the development of certain parts of the brain.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Septo-Optic Dysplasia

At least five mutations in the *HESX1* gene have been identified in people with septo-optic dysplasia. Some of these mutations change single DNA building blocks (base pairs) in the *HESX1* gene, while others insert or delete genetic material in the gene. Mutations in this gene alter the function of the HESX1 protein, for example by preventing it from binding to DNA and repressing the activity of other genes. *HESX1* gene mutations disrupt the formation and early development of the pituitary gland, optic nerves, and other brain structures. These abnormalities of brain development lead to the characteristic features of septo-optic dysplasia.

Studies suggest that mutations in the *HESX1* gene are a rare cause of septo-optic dysplasia.

### 2.2. Other Disorders

More than a dozen *HESX1* gene mutations have been found to cause underdevelopment (hypoplasia) of the pituitary gland in people without the other characteristic features of septo-optic dysplasia. Pituitary hypoplasia leads to a shortage of hormones needed for growth, reproduction, and other critical body functions. Affected individuals may also have signs and symptoms affecting other parts of the body, including genital abnormalities, vision impairment, distinctive facial features, and extra (supernumerary) fingers.

Mutations in the *HESX1* gene lead to the production of a defective or nonfunctional HESX1 protein, which disrupts the formation of the pituitary gland during critical stages of embryonic development. Some mutations prevent the HESX1 protein from binding to DNA and repressing the activity of other genes. Other mutations prevent the HESX1 protein from interacting with the PROP1 protein to coordinate brain development. It is unclear how mutations in the *HESX1* gene can cause signs and symptoms affecting other parts of the body.

## 3. Other Names for This Gene

- ANF

- CPHD5
- HESX1_HUMAN
- homeobox, ES cell expressed 1
- Rathke pouch homeobox
- RPX

## References


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