# ZEB2 Gene

Subjects: Genetics & Heredity
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Zinc finger E-box binding homeobox 2

genes

## 1. Normal Function

The *ZEB2* gene provides instructions for making a protein that plays a critical role in the formation of many organs and tissues before birth. This protein is a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of particular genes. Researchers have found that the ZEB2 protein is involved in chemical signaling pathways that regulate early growth and development.

The ZEB2 protein is active in many types of cells before birth. It appears to be particularly important for the development of the neural crest, which is a group of cells in the early embryo that give rise to many tissues and organs. Neural crest cells migrate to form portions of the nervous system, glands that produce hormones (endocrine glands), pigment cells, smooth muscle and other tissues in the heart, and many tissues in the face and skull.

The ZEB2 protein is also active in cells that are not derived from the neural crest. For example, this protein is involved in the development of the digestive tract, skeletal muscles, kidneys, and other organs.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Mowat-Wilson syndrome

More than 180 mutations in the *ZEB2* gene have been identified in people with Mowat-Wilson syndrome. These mutations almost always inactivate one copy of the *ZEB2* gene. In some cases, the entire gene is deleted. In other cases, mutations within the gene lead to the production of an abnormally short, nonfunctional version of the ZEB2 protein.

A shortage of the ZEB2 protein disrupts the formation of many organs and tissues before birth. The abnormal development of neural crest-derived structures, such as the nervous system and facial features, underlie many of the signs and symptoms of Mowat-Wilson syndrome. The role of the ZEB2 protein in the development of nerves

that control the digestive tract may help explain why many people with this condition also have Hirschsprung disease, an intestinal disorder that causes severe constipation, intestinal blockage, and enlargement of the colon.

### 2.2. Coloboma

## 3. Other Names for This Gene

- KIAA0569
- SIP-1
- SIP1
- Smad interacting-protein 1
- Smad-interacting protein 1
- SMADIP1
- ZEB2_HUMAN
- ZFHX1B
- zinc finger E-box-binding protein 2
- zinc finger homeobox 1b

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