ZIC2 Gene

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Zic family member 2

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1. Normal Function

The *ZIC2* gene provides instructions for making a protein that plays an important role in the development of the front part of the brain (forebrain). This protein is a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of certain genes. The ZIC2 protein regulates genes involved in both early and late stages of forebrain development.

2. Health Conditions Related to Genetic Changes

2.1. Nonsyndromic holoprosencephaly

More than 80 mutations in the *ZIC2* gene have been found to cause nonsyndromic holoprosencephaly. This condition occurs when the brain fails to divide into two halves (hemispheres) during early development. *ZIC2* gene mutations are the second most common cause of nonsyndromic holoprosencephaly. The facial features of individuals with *ZIC2* gene mutations are different from those with nonsyndromic holoprosencephaly caused by mutations in other genes. These distinctive facial features include a narrowing of the head at the temples, outside corners of the eyes that point upward (upslanting palpebral fissures), large ears, a short nose with upturned nostrils, and a broad and deep space between the nose and mouth (philtrum). It is unclear how mutations in the *ZIC2* gene lead to these facial features.

ZIC2 gene mutations that cause nonsyndromic holoprosencephaly reduce or eliminate the activity of the ZIC2 protein. Without enough functional ZIC2 protein, the genes involved in normal forebrain development are not properly controlled. As a result, the brain does not separate into two hemispheres. The signs and symptoms of nonsyndromic holoprosencephaly are caused by abnormal development of the brain and face.

2.2. Coloboma

3. Other Names for This Gene

- HPE5
- Zic family member 2 (odd-paired Drosophila homolog)
- Zic family member 2 (odd-paired homolog, Drosophila)
- ZIC2_HUMAN
- Zinc finger protein of the cerebellum 2
- zinc finger protein ZIC 2

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