SOX9

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SRY-box 9

genes

1. Normal Function

The *SOX9* gene provides instructions for making a protein that plays a critical role during embryonic development. The SOX9 protein is especially important for development of the skeleton and plays a key role in the determination of sex before birth. The SOX9 protein attaches (binds) to specific regions of DNA and regulates the activity of other genes, particularly those that control skeletal development and sex determination. On the basis of this action, the SOX9 protein is called a transcription factor.

2. Health Conditions Related to Genetic Changes

2.1. Campomelic dysplasia

More than 70 mutations involving the *SOX9* gene have been found to cause campomelic dysplasia, a disorder that affects skeletal development, sex determination, and other processes in the body and is often life-threatening in the newborn period. Most cases of campomelic dysplasia are caused by mutations within the *SOX9* gene. These mutations prevent the production of the SOX9 protein or result in a protein with impaired ability to function as a transcription factor. About 5 percent of cases are caused by chromosome abnormalities that occur near the *SOX9* gene. These chromosome abnormalities disrupt regions of DNA called enhancers that normally regulate the activity of the *SOX9* gene. All of these genetic changes prevent the SOX9 protein from properly controlling the genes essential for normal development of the skeleton, reproductive organs, and other parts of the body. Abnormal development of these structures causes the signs and symptoms of campomelic dysplasia.

Individuals with milder forms of campomelic dysplasia are more likely to have chromosome abnormalities near the *SOX9* gene rather than mutations within the gene.

2.2. Isolated Pierre Robin sequence

Genetic changes that occur near the *SOX9* gene cause some cases of isolated Pierre Robin sequence. Individuals with this condition have a small lower jaw (micrognathia) and a tongue that is placed further back than normal (glossoptosis), which can block the airways. Most affected individuals are also born with an opening in the roof of

the mouth (a cleft palate). These cases of Pierre Robin sequence are described as isolated because they occur without other signs and symptoms.

The genetic changes associated with isolated Pierre Robin sequence are thought to disrupt enhancer regions that normally regulate the activity of the *SOX9* gene during development of the lower jaw, which reduces *SOX9* gene activity. As a result, the SOX9 protein cannot properly control the genes essential for normal jaw development, causing micrognathia. Underdevelopment of the lower jaw affects placement of the tongue and formation of the palate, leading to glossoptosis and, often, cleft palate.

3. Other Names for This Gene

- SOX9_HUMAN
- SRA1
- SRY (sex determining region Y)-box 9
- SRY (sex-determining region Y)-box 9 protein
- SRY box 9
- transcription factor SOX9

The entry is from https://medlineplus.gov/genetics/gene/sox9

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