

# TBX5 Gene

Subjects: Genetics & Heredity

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T-box 5: The *TBX5* gene provides instructions for making a protein called T-box 5 that plays an important role in the formation of tissues and organs during embryonic development.

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## 1. Normal Function

The *TBX5* gene provides instructions for making a protein called T-box 5 that plays an important role in the formation of tissues and organs during embryonic development. This protein regulates the activity of other genes by attaching (binding) to specific regions of DNA. On the basis of this action, the T-box 5 protein is called a transcription factor.

During embryonic development, the T-box 5 protein turns on (activates) genes involved in the normal development of the hands and arms (upper limbs). The T-box 5 protein also activates genes that play an important role in the growth and development of the heart. This protein appears to be particularly important for the formation of the wall (septum) that separates the right and left sides of the heart. The T-box 5 protein is also critical to the formation of the electrical system that coordinates contractions of the heart chambers.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Holt-Oram syndrome

More than 70 mutations in the *TBX5* gene have been found to cause Holt-Oram syndrome. Most of these mutations prevent the production of the T-box 5 protein. Other mutations change one of the protein building blocks (amino acids) used to make the T-box 5 protein. Researchers believe that a change in amino acids impairs the protein's ability to bind to DNA. As a result of *TBX5* mutations, genes that are important for development of the heart and upper limbs are probably not activated. Abnormal development of the heart and upper limbs is characteristic of Holt-Oram syndrome.

## 3. Other Names for This Gene

- HOS
  - T-box transcription factor *TBX5*
  - *TBX5\_HUMAN*
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