TFAP2B Gene

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Transcription factor AP-2 beta: The TFAP2B gene provides instructions for making a protein called transcription factor AP-2β.

genes

1. Normal Function

The *TFAP2B* gene provides instructions for making a protein called transcription factor AP-2β. A transcription factor is a protein that attaches (binds) to specific regions of DNA and helps control the activity of particular genes. Transcription factor AP-2β is one of a group of related proteins called AP-2 transcription factors. These proteins regulate genes that help control cell division and the self-destruction of cells that are no longer needed (apoptosis).

Transcription factor AP-2β is involved in development before birth. In particular, this protein is active in 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. Transcription factor AP-2β also appears to play an important role in the development of the limbs.

2. Health Conditions Related to Genetic Changes

2.1. Char syndrome

Fewer than 10 mutations in the *TFAP2B* gene have been identified in people with Char syndrome. These mutations alter the structure of transcription factor AP-2β. More than half of the known mutations alter a region of the protein that is critical for DNA binding. Other mutations occur in an area of the protein that is necessary for regulating gene activity. At least two changes in the *TFAP2B* gene prevent the production of any transcription factor AP-2β. A loss of this protein's function disrupts the normal development of structures derived from the neural crest, including the heart and facial features. Abnormal development of these tissues leads to the major features of Char syndrome.

2.2. Other disorders

Studies suggest that several normal variations (polymorphisms) in the *TFAP2B* gene are associated with an increased risk of type 2 diabetes, the most common form of diabetes. People with this disease have high blood sugar levels because the body does not respond correctly to insulin, a hormone produced by the pancreas. This

hormone controls how much sugar (in the form of glucose) is passed from the bloodstream into cells to be used as energy. Researchers have proposed that polymorphisms in the *TFAP2B* gene alter cells' responsiveness to insulin, particularly fat-storing cells (adipocytes).

Although changes in the *TFAP2B* gene may be associated with type 2 diabetes, a combination of lifestyle, genetic, and environmental factors all play a part in determining the risk of this complex disorder.

3. Other Names for This Gene

- activating enhancer binding protein 2 beta
- AP-2B
- AP2-B
- AP2-beta
- AP2B_HUMAN
- MGC21381
- transcription factor AP-2 beta (activating enhancer binding protein 2 beta)

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