TBX1 Gene

Subjects: Genetics & Heredity
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T-box 1: The TBX1 gene provides instructions for making a protein called T-box 1. Genes in the T-box family play important roles in the formation of tissues and organs during embryonic development.

Keywords: genes

1. Normal Function

The *TBX1* gene provides instructions for making a protein called T-box 1. Genes in the T-box family play important roles in the formation of tissues and organs during embryonic development. To carry out these roles, proteins produced from these genes bind to specific areas of DNA. The proteins attach to critical regions near genes and help control the activity of those genes. T-box proteins are called transcription factors on the basis of this action.

The T-box 1 protein appears to be necessary for the normal development of muscles and bones of the face and neck, large arteries that carry blood out of the heart, structures in the ear, and glands such as the thymus and parathyroid. Although the T-box 1 protein acts as a transcription factor, researchers have not determined which genes are regulated by this protein.

2. Health Conditions Related to Genetic Changes

2.1. 22q11.2 deletion syndrome

Most cases of 22q11.2 deletion syndrome are caused by a deletion of a small piece of chromosome 22. This region of the chromosome contains 30 to 40 genes, including the *TBX1* gene. In a small number of affected individuals without a chromosome 22 deletion, mutations in the *TBX1* gene are thought to be responsible for the characteristic signs and symptoms of the syndrome. The identified mutations include changes in single DNA building blocks (base pairs) in the *TBX1* gene and deletions of a small amount of genetic material from the gene. Some of these mutations reduce the amount of T-box 1 protein that is produced in cells, while other mutations alter the protein's function. These genetic changes likely affect the ability of the T-box 1 protein to bind to DNA and regulate the activity of other genes.

Researchers believe that changes in the *TBX1* gene, due to either a mutation in the gene or a deletion of part of chromosome 22, are responsible for many of the features of 22q11.2 deletion syndrome. Specifically, a reduction in the amount of T-box 1 or changes in the protein's normal function are associated with heart defects, an opening in the roof of the mouth (a cleft palate), distinctive facial features, hearing loss, and low calcium levels. Some studies suggest that a loss of the *TBX1* gene may also be associated with behavioral problems in affected individuals.

3. Other Names for This Gene

- CAFS
- CTHM
- DGCR
- DGS
- DORV
- TBX1_HUMAN
- TBX1C
- Testis-specific T-box protein
- TGA
- VCFS

References

1. Arnold JS, Braunstein EM, Ohyama T, Groves AK, Adams JC, Brown MC, Morrow BE. Tissue-specific roles of Tbx1 in the development of the outer, middle and innerear, defective in 22q11DS patients. Hum Mol Genet. 2006 May 15;15(10):1629-39.
2. Baldini A. DiGeorge syndrome: an update. Curr Opin Cardiol. 2004May;19(3):201-4. Review.
3. Baldini A. DiGeorge's syndrome: a gene at last. Lancet. 2003 Oct25;362(9393):1342-3.
4. Baldini A. Dissecting contiguous gene defects: TBX1. Curr Opin Genet Dev. 2005Jun;15(3):279-84. Review.
5. Packham EA, Brook JD. T-box genes in human disorders. Hum Mol Genet. 2003 Apr 1;12 Spec No 1:R37-44. Review.
6. Paylor R, Glaser B, Mupo A, Ataliotis P, Spencer C, Sobotka A, Sparks C, Choi CH, Oghalai J, Curran S, Murphy KC, Monks S, Williams N, O'Donovan MC, Owen MJ,Scambler PJ, Lindsay E. Tbx1 haploinsufficiency is linked to behavioral disordersin mice and humans: implications for 22q11 deletion syndrome. Proc Natl Acad Sci U S A. 2006 May 16;103(20):7729-34.
7. Plageman TF Jr, Yutzey KE. T-box genes and heart development: putting the "T" in heart. Dev Dyn. 2005 Jan;232(1):11-20. Review.
8. Stoller JZ, Epstein JA. Identification of a novel nuclear localization signal in Tbx1 that is deleted in DiGeorge syndrome patients harboring the 1223delCmutation. Hum Mol Genet. 2005 Apr 1;14(7):885-92.
9. Weksberg R, Stachon AC, Squire JA, Moldovan L, Bayani J, Meyn S, Chow E,Bassett AS. Molecular characterization of deletion breakpoints in adults with22q11 deletion syndrome. Hum Genet. 2007 Feb;120(6):837-45.
10. Yagi H, Furutani Y, Hamada H, Sasaki T, Asakawa S, Minoshima S, Ichida F, Joo K, Kimura M, Imamura S, Kamatani N, Momma K, Takao A, Nakazawa M, Shimizu N,Matsuoka R. Role of TBX1 in human del22q11.2 syndrome. Lancet. 2003 Oct25;362(9393):1366-73.
11. Yamagishi H, Srivastava D. Unraveling the genetic and developmental mysteries of 22q11 deletion syndrome. Trends Mol Med. 2003 Sep;9(9):383-9. Review.
12. Zweier C, Sticht H, Aydin-Yaylagül I, Campbell CE, Rauch A. Human TBX1missense mutations cause gain of function resulting in the same phenotype as22q11.2 deletions. Am J Hum Genet. 2007 Mar;80(3):510-7.
