TBXT gene

T-box transcription factor T

Normal Function

The *TBXT* gene provides instructions for making a protein called brachyury. Brachyury is a member of a protein family called T-box proteins, which play critical roles during embryonic development. T-box proteins regulate the activity of other genes by attaching (binding) to specific regions of DNA. On the basis of this action, T-box proteins are called transcription factors.

The brachyury protein is important for the development of the notochord, which is the precursor of the spinal column in the embryo. The notochord disappears before birth, but in a small percentage of individuals, some of its cells remain in the base of the skull or in the spine. The notochord helps control the development of the neural tube, which is a layer of cells that ultimately develops into the brain and spinal cord.

Health Conditions Related to Genetic Changes

Chordoma

Changes in the *TBXT* gene have been associated with chordoma. A chordoma is a cancerous tumor that occurs along the spine or in the base of the skull, gradually extending further into the bone and into nearby soft tissue. The tumor develops from residual cells of the notochord that begin to grow and divide uncontrollably.

An inherited duplication of the *TBXT* gene identified in a few families is associated with an increased risk of developing a chordoma. Duplications or increases in activity (expression) of the *TBXT* gene have also been identified in people with chordoma who have no history of the disorder in their family. In these individuals, the changes occur only in the tumor cells and are not inherited.

Duplications and increases in expression of the *TBXT* gene both result in the production of excess brachyury protein. The specific mechanism by which excess brachyury protein contributes to the development of chordomas is unclear.

Other disorders

A variation of the *TBXT* gene is associated with an increased risk of neural tube defects, including spina bifida. Neural tube defects, which can result in spine and brain

abnormalities of varying severity, result when the neural tube fails to close completely during the first few weeks of embryonic development.

The *TBXT* gene variation associated with neural tube defects, known as TIVS7 T/C, changes a single DNA building block (nucleotide) in a region of the gene known as intron 7. The effect of this genetic change is unclear. Researchers suggest that it may affect the regulation of brachyury protein production in early development, increasing the risk of neural tube defects.

Other Names for This Gene

- BRAC_HUMAN
- brachyury protein
- protein T
- T
- T brachyury homolog
- T brachyury transcription factor
- T, brachyury homolog (mouse)
- TFT

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TBXT (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6862[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28brachyury%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- T-BOX TRANSCRIPTION FACTOR T; TBXT (https://omim.org/entry/601397)
- NEURAL TUBE DEFECTS, SUSCEPTIBILITY TO; NTD (https://omim.org/entry/182940)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6862)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TBXT[gene])

References

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Genomic Location

The *TBXT* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

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