# TGIF1 gene

TGFB induced factor homeobox 1

## Normal Function

The *TGIF1* gene provides instructions for making a protein called TG-interacting factor. This protein is important for normal development of the front part of the brain (forebrain). TG-interacting factor is a transcription factor, which means that it regulates the activity of certain genes. This protein turns off genes by attaching (binding) to specific regions of DNA or by interacting with other DNA-binding proteins.

TG-interacting factor regulates signaling pathways that are important for embryonic development. This protein blocks the signals of the transforming growth factor beta ( TGF-β) pathway. This signaling pathway transmits chemical signals from the cell surface to the nucleus, which allows the environment outside the cell to affect how the cell produces other proteins. TG-interacting factor also blocks a molecule called retinoic acid from regulating gene activity. Retinoic acid, a form of vitamin A, binds to a group of transcription factors that regulate a number of genes important for early development. By blocking these signaling pathways, TG-interacting factor ensures that certain genes are turned off at the proper time.

## Health Conditions Related to Genetic Changes

Nonsyndromic holoprosencephaly

At least 13 mutations in the *TGIF1* gene have been found to cause nonsyndromic holoprosencephaly. This condition occurs when the brain fails to divide into two halves ( hemispheres) during early development. *TGIF1* gene mutations are the fourth most common cause of nonsyndromic holoprosencephaly. These mutations disrupt the protein's ability to bind with DNA or interact with other proteins. As a result, TG-interacting factor cannot block the signals of the TGF-β pathway and retinoic acid. If the signals involved in forebrain development are not properly regulated, the brain does not separate into two hemispheres. The signs and symptoms of nonsyndromic holoprosencephaly are caused by abnormal development of the brain and face.

## Other Names for This Gene

- 5'-TG-3' interacting factor
- 5'-TG-3'-interacting factor 1

- homeobox protein TGIF
- homeobox protein TGIF1
- HPE4
- MGC39747
- MGC5066
- TALE homeobox TG-interacting factor
- TGFB-induced factor homeobox 1
- TGIF
- TGIF1_HUMAN
- transforming growth factor-beta-induced factor

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TGIF1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7050[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28TGIF1%5BTIAB%5D%29+OR+%28%28HPE4%5BTIAB%5D%29+OR+%28TGIF%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TRANSFORMING GROWTH FACTOR-BETA-INDUCED FACTOR; TGIF (https://omim.org/entry/602630)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7050)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TGIF1[gene])

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## Genomic Location

The *TGIF1* gene is found on chromosome 18 (https://medlineplus.gov/genetics/chromosome/18/).

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