



## TBXAS1 gene

thromboxane A synthase 1

### Normal Function

The *TBXAS1* gene provides instructions for making an enzyme called thromboxane A synthase 1. This enzyme acts as part of a chemical pathway called the arachidonic acid cascade. Through this multistep pathway, a molecule called arachidonic acid is processed to produce several molecules with diverse functions in the body. As part of this pathway, thromboxane A synthase 1 converts a molecule called prostaglandin H<sub>2</sub> into another molecule called thromboxane A<sub>2</sub>. Thromboxane A<sub>2</sub> is involved in normal blood clotting (hemostasis), playing critical roles in the narrowing of blood vessels (vasoconstriction) to slow blood flow and the clumping (aggregation) of blood cell fragments called platelets at the site of an injury.

Studies suggest that the activity of thromboxane A synthase 1 may also be important for bone remodeling, which is a normal process in which old bone is removed and new bone is created to replace it, and for the production of red blood cells in bone marrow.

### Health Conditions Related to Genetic Changes

#### Ghosal hematodiaphyseal dysplasia

At least four mutations in the *TBXAS1* gene have been found to cause Ghosal hematodiaphyseal dysplasia. This condition is characterized by abnormally thick bones and a shortage of red blood cells (anemia) caused by scarring (fibrosis) of the bone marrow.

Each of the known mutations changes a single protein building block (amino acid) in thromboxane A synthase 1, which severely reduces the activity of the enzyme. A shortage of this enzyme's activity prevents the conversion of prostaglandin H<sub>2</sub> to thromboxane A<sub>2</sub>. As a result, cells have more prostaglandin H<sub>2</sub> than usual. Prostaglandin H<sub>2</sub> is converted into several related molecules, including prostaglandin E<sub>2</sub>, which is thought to be involved in bone remodeling and in controlling the growth of immature red blood cells. Researchers speculate that an increase in prostaglandin E<sub>2</sub> levels resulting from excess prostaglandin H<sub>2</sub> contributes to the bone abnormalities and anemia that occur in people with Ghosal hematodiaphyseal dysplasia. However, the exact mechanism by which a lack of thromboxane A synthase 1 activity leads to the particular features of this condition is still unclear.

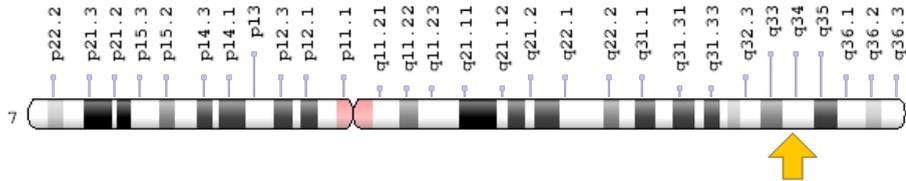
A shortage of thromboxane A synthase 1 activity also reduces the level of thromboxane A<sub>2</sub> in cells. Although this molecule plays a critical role in hemostasis, people with Ghosal hematodiaphyseal dysplasia do not appear to have problems with

blood clotting. Researchers suspect that other molecules involved in vasoconstriction and platelet aggregation may be able to compensate for the lack of thromboxane A<sub>2</sub> in these individuals.

### Chromosomal Location

Cytogenetic Location: 7q34, which is the long (q) arm of chromosome 7 at position 34

Molecular Location: base pairs 139,778,242 to 140,020,321 on chromosome 7 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- BDPLT14
- CYP5
- CYP5A1
- cytochrome P450 5A1
- cytochrome P450, family 5, subfamily A, polypeptide 1
- GHOSAL
- platelet, cytochrome P450, subfamily V
- THAS
- THAS\_HUMAN
- thromboxane-A synthase
- thromboxane A synthase 1 (platelet)
- thromboxane A synthase 1 (platelet, cytochrome P450, family 5, subfamily A)
- TS
- TXA synthase
- TXAS
- TXS

## Additional Information & Resources

### Educational Resources

- Platelet Research Laboratory: Platelet Function  
[http://www.platelet-research.org/1/function\\_acti.htm](http://www.platelet-research.org/1/function_acti.htm)

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TBXAS1%5BTIAB%5D%29+OR+%28thromboxane+A+synthase%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

### Catalog of Genes and Diseases from OMIM

- THROMBOXANE A SYNTHASE 1  
<http://omim.org/entry/274180>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_TBXAS1.html](http://atlasgeneticsoncology.org/Genes/GC_TBXAS1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=TBXAS1%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:11609](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:11609)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:6916>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6916>
- UniProt  
<https://www.uniprot.org/uniprot/P24557>

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