

## TG gene

thyroglobulin

### Normal Function

The *TG* gene provides instructions for making a protein called thyroglobulin, one of the largest proteins in the body. This protein is found only in the thyroid gland, a butterfly-shaped tissue in the lower neck. Thyroglobulin combines with iodine and is modified and broken down to release small molecules known as thyroid hormones. Thyroid hormones play an important role in regulating growth, brain development, and the rate of chemical reactions in the body (metabolism). Thyroglobulin also serves as a protein storehouse for iodine and inactive thyroid hormone until these substances are needed.

### Health Conditions Related to Genetic Changes

#### Congenital hypothyroidism

Mutations in the *TG* gene can cause congenital hypothyroidism, a condition characterized by abnormally low levels of thyroid hormones starting from birth. The *TG* gene mutations involved in this condition either delete a small segment of the *TG* gene or change one of the DNA building blocks (base pairs). As a result, the 3-dimensional shape of thyroglobulin is altered, reducing the amount of properly structured protein that is available for thyroid hormone production. In most affected individuals, the thyroid gland is enlarged (goiter) in an attempt to compensate for reduced hormone production. Because cases caused by *TG* gene mutations are due to a disruption of thyroid hormone synthesis, they are classified as thyroid dysmorphogenesis.

#### Graves's disease

MedlinePlus Genetics provides information about Graves's disease

#### Hashimoto's disease

MedlinePlus Genetics provides information about Hashimoto's disease

#### Other disorders

*TG* gene mutations have also been identified in some people who have a goiter but normal or near normal thyroglobulin levels. These mutations either delete part of the *TG*

gene or change one of the DNA base pairs. As a result, the 3-dimensional shape of thyroglobulin is altered, reducing the amount of properly structured protein that is available for thyroid hormone production. The thyroid gland enlarges, forming a goiter, to compensate for decreased levels of thyroglobulin. With this compensation, thyroid hormone levels are normal, so affected individuals do not have other signs and symptoms of congenital hypothyroidism (described above). It is unclear why enlargement of the thyroid gland can compensate in some affected individuals but not others (leading to congenital hypothyroidism).

### Other Names for This Gene

- AITD3
- TGN
- THYG\_HUMAN

### Additional Information & Resources

#### Tests Listed in the Genetic Testing Registry

- Tests of TG ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7038\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7038[geneid]))

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TG%5BTIAB%5D%29+OR+%28thyroglobulin%5BTIAB%5D%29%29+AND+%28thyroglobulin%5BMAJR%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29%29>)

#### Catalog of Genes and Diseases from OMIM

- THYROGLOBULIN; TG (<https://omim.org/entry/188450>)

#### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7038>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=TG\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=TG[gene]))

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## Genomic Location

The *TG* gene is found on chromosome 8 (<https://medlineplus.gov/genetics/chromosome>)

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