DUOX2 gene

dual oxidase 2

Normal Function

The *DUOX2* gene provides instructions for making an enzyme called dual oxidase 2. This enzyme is found in the thyroid gland, which is a butterfly-shaped tissue in the lower neck. The enzyme is also found in salivary glands, the digestive tract, and airways in the throat and lungs. Dual oxidase 2 helps generate a chemical called hydrogen peroxide. In the thyroid, hydrogen peroxide is required for one of the final steps in the production of thyroid hormones. Thyroid hormones play an important role in regulating growth, brain development, and the rate of chemical reactions in the body (metabolism).

Health Conditions Related to Genetic Changes

Congenital hypothyroidism

Researchers have identified several *DUOX2* gene mutations that cause congenital hypothyroidism, a condition characterized by a reduction of thyroid hormone levels that is present from birth. Most of these mutations result in an abnormally small version of the dual oxidase 2 enzyme. The remaining mutations change one of the building blocks (amino acids) used to make the enzyme, which probably alters the enzyme's structure. All *DUOX2* gene mutations limit the enzyme's ability to generate hydrogen peroxide. Without sufficient hydrogen peroxide, thyroid hormone production is disrupted. In some cases, the thyroid gland is enlarged (goiter) in an attempt to compensate for reduced thyroid hormone production. Because cases caused by mutations in the *DUOX2* gene are due to a disruption of thyroid hormone synthesis, they are classified as thyroid dyshormonogenesis.

The reduction in thyroid hormone production is affected by the number of *DUOX2* genes with a mutation. Each cell in the body has two copies of the *DUOX2* gene. If both copies of the gene have a mutation, cells in the thyroid gland generate very little hydrogen peroxide. As a result, thyroid hormone levels are extremely low, causing severe congenital hypothyroidism. If only one copy of the *DUOX2* gene is mutated, some hydrogen peroxide is produced. As a result, thyroid hormone levels are slightly reduced, causing mild congenital hypothyroidism. Sometimes, mild congenital hypothyroidism is temporary (transient), and thyroid hormone levels that are low during infancy increase with age.

Other Names for This Gene

- DUOX2_HUMAN
- flavoprotein NADPH oxidase
- LNOX2
- NADPH thyroid oxidase 2
- nicotinamide adenine dinucleotide phosphate oxidase
- NOXEF2
- P138-TOX
- THOX2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DUOX2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=50506[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DUOX2%5BTIAB%5D%29+OR+%28dual+oxidase+2%5BTIAB%5D%29%29+OR+%28%28dual+oxidase+2+precursor%5BTIAB%5D%29+OR+%28LNOX2%5BTIAB%5D%29+OR+%28THOX2%5BTIAB%5D%29+OR+%28P138-TOX%5BTIAB%5D%29+OR+%28NADPH+thyroid+oxidase+2%5BTIAB%5D%29+OR+%28dual+oxidase-like+domains+2%5BTIAB%5D%29+OR+%28NADPH+oxidase/peroxidase+DUOX2%5BTIAB%5D%29+OR+%28NADH/NADPH+thyroid+oxidase+p138-tox%5BTIAB%5D%29+OR+%28nicotinamide+adenine+dinucleotide+phosphate+oxidase%5BTIAB%5D%29+OR+%28P138%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- DUAL OXIDASE 2; DUOX2 (https://omim.org/entry/606759)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/50506)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DUOX2[gene])

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Genomic Location

The *DUOX2* gene is found on chromosome 15 (https://medlineplus.gov/genetics/chromosome/15/).

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