



FOXP3 gene

forkhead box P3

Normal Function

The *FOXP3* gene provides instructions for producing the forkhead box P3 (FOXP3) protein. The FOXP3 protein attaches (binds) to specific regions of DNA and helps control the activity of genes that are involved in regulating the immune system. The immune system normally protects the body from foreign invaders, such as bacteria and viruses, by recognizing and attacking these invaders and clearing them from the body.

On the basis of its role in controlling gene activity, the FOXP3 protein is called a transcription factor. This protein is essential for the production and normal function of certain immune cells called regulatory T cells, which play an important role in preventing autoimmunity. Autoimmunity occurs when the body attacks its own tissues and organs by mistake. The FOXP3 protein is found primarily in an immune system gland called the thymus, where these regulatory T cells are produced.

Health Conditions Related to Genetic Changes

Immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome

More than 60 mutations in the *FOXP3* gene have been found to cause immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome. This rare condition is characterized by the development of multiple autoimmune disorders in affected individuals, typically affecting the intestines, skin, and hormone-producing (endocrine) glands. Most of the *FOXP3* gene mutations involved in IPEX syndrome change a protein building block (amino acid) in the region of the FOXP3 protein that binds to DNA or lead to the production of an abnormally short, nonfunctional protein. Mutations in the *FOXP3* gene impair the normal function of regulatory T cells. Without the function of these cells, the body cannot control immune responses. Normal body tissues and organs are attacked, causing the multiple autoimmune disorders that develop in people with IPEX syndrome.

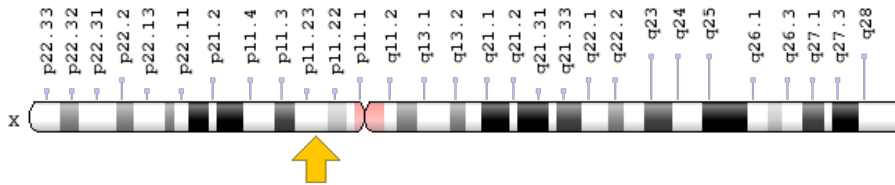
Hashimoto thyroiditis

Type 1 diabetes

Chromosomal Location

Cytogenetic Location: Xp11.23, which is the short (p) arm of the X chromosome at position 11.23

Molecular Location: base pairs 49,250,436 to 49,264,932 on the X chromosome (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AIID
- DIETER
- FOXP3_HUMAN
- immune dysregulation, polyendocrinopathy, enteropathy, X-linked
- immunodeficiency, polyendocrinopathy, enteropathy, X-linked
- IPEX
- JM2
- MGC141961
- MGC141963
- PIDX
- scurfin
- XPID

Additional Information & Resources

Educational Resources

- Immunobiology (fifth edition, 2001): Autoimmune Responses are Directed Against Self Antigens
<https://www.ncbi.nlm.nih.gov/books/NBK27155/>

Clinical Information from GeneReviews

- IPEX Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1118>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28FOXP3%5BTIAB%5D%29+OR+%28forkhead+box+P3%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+360+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- FORKHEAD BOX P3
<http://omim.org/entry/300292>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/FOXP3ID44129chXp11.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=FOXP3%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6106
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:50943>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/50943>
- UniProt
<https://www.uniprot.org/uniprot/Q9BZS1>

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