ZAP70 gene
zeta chain of T cell receptor associated protein kinase 70

Normal Function

The ZAP70 gene provides instructions for making a protein called zeta-chain-associated protein kinase. This protein is part of a signaling pathway that directs the development of and turns on (activates) immune system cells called T cells. T cells identify foreign substances and defend the body against infection.

The ZAP70 gene is important for the development and function of several types of T cells. These include cytotoxic T cells (CD8+ T cells), whose functions include destroying cells infected by viruses. The ZAP70 gene is also involved in the activation of helper T cells (CD4+ T cells). These cells direct and assist the functions of the immune system by influencing the activities of other immune system cells.

Health Conditions Related to Genetic Changes

**ZAP70-related severe combined immunodeficiency**

More than 12 mutations in the ZAP70 gene have been identified in people with ZAP70-related severe combined immunodeficiency (SCID). These mutations either change single protein building blocks (amino acids) in the protein sequence or disrupt how genetic information is pieced together to make the blueprint for producing the protein.

Mutations in the ZAP70 gene prevent the production of zeta-chain-associated protein kinase or result in a protein that is unstable and cannot perform its function. A loss of functional zeta-chain-associated protein kinase leads to the absence of CD8+ T cells and an excess of inactive CD4+ T cells. The resulting shortage of active T cells causes people with ZAP70-related SCID to be more susceptible to infection.
Chromosomal Location

Cytogenetic Location: 2q11.2, which is the long (q) arm of chromosome 2 at position 11.2

Molecular Location: base pairs 97,712,030 to 97,744,327 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FLJ17670
- FLJ17679
- SRK
- STD
- syk-related tyrosine kinase
- TZK
- ZAP-70
- ZAP70_HUMAN
- zeta-chain (TCR) associated protein kinase 70kDa
- zeta-chain associated protein kinase 70kDa
- zeta-chain associated protein kinase, 70kD
- zeta chain of T-cell receptor associated protein kinase 70
- zeta chain of T cell receptor associated protein kinase 70kDa

Additional Information & Resources

Educational Resources

  https://www.ncbi.nlm.nih.gov/books/NBK27109/#A1509
Clinical Information from GeneReviews

- ZAP70-Related Combined Immunodeficiency
  https://www.ncbi.nlm.nih.gov/books/NBK20221

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ZAP70%5BTIAB%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

Catalog of Genes and Diseases from OMIM

- ZETA-CHAIN-ASSOCIATED PROTEIN KINASE
  http://omim.org/entry/176947

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/ZAP70ID197ch2q11.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ZAP70%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:7535
- Mutation Registry for ZAP70 Deficiency
  http://structure.bmc.lu.se/idbase/ZAP70base/index.php
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P43403

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9407944
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