

# ZAP70 Gene

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Contributor: Peter Tang

Zeta chain of T cell receptor associated protein kinase 70

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## 1. 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.

## 2. Health Conditions Related to Genetic Changes

### 2.1. 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.

## 3. Other Names for This Gene

- FLJ17670
- FLJ17679
- SRK
- STD
- syk-related tyrosine kinase
- TZK
- ZAP-70
- ZAP70\_HUMAN
- zeta chain of T cell receptor associated protein kinase 70kDa
- zeta chain of T-cell receptor associated protein kinase 70
- zeta-chain (TCR) associated protein kinase 70kDa

- zeta-chain associated protein kinase 70kDa
- zeta-chain associated protein kinase, 70kD

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