

## TAP1 gene

transporter 1, ATP binding cassette subfamily B member

### Normal Function

The *TAP1* gene provides instructions for making a protein that plays an important role in the immune system. The TAP1 protein assembles with another protein called TAP2 (produced from the *TAP2* gene) to form a protein complex called transporter associated with antigen processing (TAP) complex. This complex, which is found in the membrane of a cell structure called the endoplasmic reticulum, moves (transports) protein fragments (peptides) from foreign invaders into the endoplasmic reticulum. There, the peptides are attached to major histocompatibility complex (MHC) class I proteins. The peptide-bound MHC class I proteins are then moved to the surface of the cell so that specialized immune system cells can interact with them. When these immune system cells recognize the peptides as harmful, they launch an immune response to get rid of the foreign invaders.

### Health Conditions Related to Genetic Changes

#### Bare lymphocyte syndrome type I

At least six mutations in the *TAP1* gene have been found to cause bare lymphocyte syndrome type I (BLS I). This immune system disorder often causes recurrent bacterial infections in the respiratory tract and open sores (ulcers) on the skin, although some people with BLS I have no symptoms of the condition. *TAP1* gene mutations involved in BLS I prevent production of functional TAP1 protein. Absence of functional TAP1 impairs the formation of the TAP complex, without which peptides from foreign invaders cannot be transported into the endoplasmic reticulum and attached to MHC class I proteins. Consequently, MHC class I proteins are broken down, which results in a shortage of these proteins on the surface of cells. A lack of MHC class I proteins impairs the body's immune response to bacteria, leading to recurrent bacterial infections. Researchers are unsure why people with BLS I do not also get viral infections, but they suspect that other immune processes are able to recognize and fight viruses. It is also not clear how *TAP1* gene mutations are involved in the development of skin ulcers.

### Other Names for This Gene

- ABC transporter, MHC 1
- ABC17

- ABCB2
- APT1
- ATP-binding cassette sub-family B member 2
- ATP-binding cassette, sub-family B (MDR/TAP), member 2
- D6S114E
- peptide supply factor 1
- peptide transporter involved in antigen processing 1
- peptide transporter PSF1
- peptide transporter TAP1
- PSF-1
- PSF1
- RING4
- TAP1N
- transporter 1 ATP-binding cassette sub-family B
- transporter 1, ATP-binding cassette, sub-family B (MDR/TAP)
- transporter associated with antigen processing
- transporter, ATP-binding cassette, major histocompatibility complex, 1

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of TAP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6890\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6890[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TAP1%5BTIAB%5D%29+OR+%28transporter+1,+ATP+binding+cassette+subfamily+B+member%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+1800+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- TRANSPORTER, ATP-BINDING CASSETTE, MAJOR HISTOCOMPATIBILITY COMPLEX, 1; TAP1 (<https://omim.org/entry/170260>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6890>)

- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=TAP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=TAP1[gene]))

## References

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## Genomic Location

The *TAP1* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

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