

ERAP1 gene

endoplasmic reticulum aminopeptidase 1

Normal Function

The *ERAP1* gene provides instructions for making a protein called endoplasmic reticulum aminopeptidase 1. As its name suggests, this protein is active in a cellular structure called the endoplasmic reticulum, which is involved in protein processing and transport. This protein is an aminopeptidase, which is an enzyme that cuts (cleaves) other proteins into smaller fragments called peptides.

Endoplasmic reticulum aminopeptidase 1 has two major functions, both of which are important for normal immune system function. First, the protein cleaves several other proteins called cytokine receptors on the surface of cells. Cleaving these receptors reduces their ability to transmit chemical signals into the cell, which affects the process of inflammation.

Second, endoplasmic reticulum aminopeptidase 1 cleaves many types of proteins into small peptides that can be recognized by the immune system. These peptides are exported to the cell surface, where they attach to major histocompatibility complex (MHC) class I proteins. MHC class I proteins display the peptides to the immune system. If the immune system recognizes the peptides as foreign (such as viral or bacterial peptides), it responds by triggering the infected cell to self-destruct.

While the protein is involved in the normal functioning of the immune system, it plays a particular role in protecting the body against the development of autoimmune disorders and cancer.

Health Conditions Related to Genetic Changes

Ankylosing spondylitis

Variations (polymorphisms) in the *ERAP1* gene have been found to influence the risk of ankylosing spondylitis. This condition is a form of painful, ongoing joint inflammation (chronic inflammatory arthritis) that primarily affects the spine. Each of the *ERAP1* gene variants changes a single protein building block (amino acid) in endoplasmic reticulum aminopeptidase 1. Individuals with an *ERAP1* polymorphism who go on to develop ankylosing spondylitis tend to also have a variant in the *HLA-B* gene (specifically, the *HLA-B27* variant).

Researchers believe that changes in the structure of endoplasmic reticulum aminopeptidase 1 could alter either of its two major functions. Other genetic and environmental factors, many of which are unknown, also affect the chance of developing ankylosing spondylitis.

Other Names for This Gene

- A-LAP
- adipocyte-derived leucine aminopeptidase
- ALAP
- aminopeptidase PILS
- aminopeptidase regulator of TNFR1 shedding
- APPILS
- ARTS-1
- ARTS1
- ERAAP
- ERAAP1
- ERAP1_HUMAN
- KIAA0525
- PILS-AP
- PILSAP
- puromycin-insensitive leucyl-specific aminopeptidase
- type 1 tumor necrosis factor receptor shedding aminopeptidase regulator

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ERAP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51752\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51752[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ERAP1%5BTIAB%5D%29+OR+%28endoplasmic+reticulum+aminopeptidase+1%5BTIAB%5D%29%29+OR+%28%28ARTS-1%5BTIAB%5D%29+OR+%28ARTS1%5BTIAB%5D%29+OR+%28ERAAP%5BTIAB%5D%29+OR+%28PILS-AP%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%29>)

Catalog of Genes and Diseases from OMIM

- ENDOPLASMIC RETICULUM AMINOPEPTIDASE 1; ERAP1 (<https://omim.org/entry/606832>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/51752>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ERAP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ERAP1[gene]))

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Genomic Location

The *ERAP1* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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