IL1A gene

interleukin 1 alpha

Normal Function

The *IL1A* gene provides instructions for making a protein called interleukin-1 alpha. Interleukins are a group of proteins that are made primarily in immune system cells. They are involved in cell-to-cell communication and have a wide variety of functions within the immune system. Interleukin-1 alpha is described as "pro-inflammatory" because it stimulates the activity of genes involved in inflammation and immunity. This protein plays a critical role in protecting the body from foreign invaders such as bacteria and viruses. It is also involved in bone resorption, the breakdown and removal of bone tissue that is no longer needed.

Interleukin-1 alpha is initially produced as a relatively long protein that is trapped within cells. Another protein, called calpain, cuts (cleaves) this precursor protein to create a shorter, mature version of interleukin-1 alpha. The shorter form of this protein is secreted by immune system cells to influence the functions of other cells.

Health Conditions Related to Genetic Changes

Ankylosing spondylitis

Several variations (polymorphisms) in the *IL1A* 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 these variations changes a single protein building block (amino acid) in interleukin-1 alpha. It is not fully known how these variations alter the protein's function. Studies suggest that the effects of *IL1A* variations are probably related to the role of interleukin-1 alpha in promoting inflammation. Other genetic and environmental factors, many of which are unknown, also affect the chance of developing ankylosing spondylitis.

Idiopathic inflammatory myopathy

MedlinePlus Genetics provides information about Idiopathic inflammatory myopathy

Intervertebral disc disease

MedlinePlus Genetics provides information about Intervertebral disc disease

Keratoconus

MedlinePlus Genetics provides information about Keratoconus

Other disorders

Variations in the *IL1A* gene have been studied as potential risk factors for several other disorders associated with abnormal inflammation. These include chronic gum (periodontal) disease, a progressive bone infection known as chronic osteomyelitis, and an eye disease called open-angle glaucoma.

The most well-studied variation affecting the *IL1A* gene is a change in a single DNA building block (nucleotide) in a region of regulatory DNA near the start of the gene (written as IL1A-889 C>T). This variation affects the production of interleukin-1 alpha within cells. Researchers have also identified a second common variation in the *IL1A* gene, written as IL1A+4845 G>T, which changes a single nucleotide in the gene. This variation likely affects the sensitivity of interleukin-1 alpha to cleavage by calpain.

It is unclear how changes in the *IL1A* gene influence the risk of inflammatory disorders. Studies suggest that the effects of *IL1A* variations are probably related to the role of interleukin-1 alpha in promoting inflammation. Other genetic and environmental factors also likely affect the chance of developing these complex disorders.

Other Names for This Gene

- hematopoietin-1
- IL-1 alpha
- IL-1A
- IL1
- IL1-ALPHA
- IL1A_HUMAN
- IL1F1
- interleukin 1, alpha
- Interleukin-1 alpha
- preinterleukin 1 alpha
- pro-interleukin-1-alpha

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of IL1A (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3552[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28IL1A%5BTI%5D%29+OR+%28IL-1+alpha%5BTI%5D%29+OR+%28IL-1A%5BTI%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- INTERLEUKIN 1-ALPHA; IL1A (https://omim.org/entry/147760)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3552)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=IL1A[gene])

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Genomic Location

The *IL1A* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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