

ADA2 gene

adenosine deaminase 2

Normal Function

The *ADA2* gene provides instructions for making an enzyme called adenosine deaminase 2. This enzyme breaks down molecules called adenosine and 2'-deoxyadenosine. Because this enzyme functions in the spaces between cells, it is described as extracellular. Another form of the enzyme, adenosine deaminase 1, breaks down the same molecules inside cells. This other version of the enzyme is produced from the *ADA* gene.

Researchers are still working to determine the functions of adenosine deaminase 2. Studies suggest that it acts as a growth factor, which means that it stimulates cell growth and division. In particular, the enzyme appears to be involved in the growth and development of certain immune system cells, including macrophages, which are a type of white blood cell that plays a critical role in inflammation. Inflammation is a normal immune system response to injury and foreign invaders (such as bacteria). Some macrophages are pro-inflammatory, meaning they promote inflammation, while others are anti-inflammatory, meaning they reduce inflammation.

Health Conditions Related to Genetic Changes

Adenosine deaminase 2 deficiency

More than 60 mutations in the *ADA2* gene have been found to cause adenosine deaminase 2 deficiency, a disorder characterized by abnormal inflammation of various organs and tissues, particularly the blood vessels (vasculitis). These mutations severely reduce or eliminate the function of adenosine deaminase 2. Researchers do not fully understand how a shortage (deficiency) of this enzyme's activity leads to vasculitis and immune system abnormalities. They speculate that the enzyme deficiency may disrupt the balance between pro-inflammatory and anti-inflammatory macrophages in various tissues, leading to a buildup of pro-inflammatory macrophages and abnormal inflammation.

Other Names for This Gene

- adenosine deaminase CECR1
- ADGF

- cat eye syndrome chromosome region, candidate 1
- cat eye syndrome critical region protein 1
- CECR1
- IDGFL
- SNEDS

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ADA2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51816\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51816[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28CECR1%5BTIAB%5D%29+OR+%28adenosine+deaminase+2%5BTIAB%5D%29+OR+%28ADA2%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- ADENOSINE DEAMINASE 2; ADA2 (<https://omim.org/entry/607575>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/51816>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ADA2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ADA2[gene]))

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Genomic Location

The *ADA2* gene is found on chromosome 22 (<https://medlineplus.gov/genetics/chromosome/22/>).

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