

ADAR gene

adenosine deaminase RNA specific

Normal Function

The *ADAR* gene provides instructions for making a protein called RNA-specific adenosine deaminase 1 (ADAR1). This protein is involved in making changes to (editing) ribonucleic acid (RNA), a chemical cousin of DNA. Specifically, it attaches (binds) to RNA and changes an RNA building block (nucleotide) called adenosine to another nucleotide called inosine.

The ADAR1 protein is involved in the control of the innate immune response, which is the immune system's early response to foreign invaders (pathogens). The adenosine-to-inosine editing performed by ADAR1 is thought to change certain areas of the body's own RNA that the immune system might interpret as belonging to a virus that should be attacked. In this way, the protein helps the immune system avoid inappropriate targeting of the body's own tissues.

The ADAR1 protein is also thought to inhibit the replication and spread of certain viruses, such as human immunodeficiency virus (HIV) and hepatitis C, by modifying their RNA. In addition, the ADAR1 protein controls the function of certain chemical messengers called neurotransmitters at particular sites in the body by modifying the RNA blueprint for receptor proteins that interact with the neurotransmitters. Studies suggest that the ADAR1 protein may have other functions that are not well understood.

Health Conditions Related to Genetic Changes

Aicardi-Goutières syndrome

At least 30 *ADAR* gene mutations have been identified in people with Aicardi-Goutières syndrome, a disorder that involves severe brain dysfunction (encephalopathy), skin lesions, immune system abnormalities, and other health problems. Some of these mutations lead to an ADAR1 protein that is less able to bind to RNA; others impair the protein's RNA editing function. As a result, control of the immune response is impaired and the immune system attacks the body's own tissues and organs, leading to the signs and symptoms of Aicardi-Goutières syndrome.

Other disorders

More than 180 *ADAR* gene mutations have been identified in people with dyschromatosis symmetrica hereditaria. This disorder is characterized by freckle-like spots (macules) on the face, hands, and feet that are darker or lighter than surrounding skin, generally appearing in infancy or early childhood.

The *ADAR* gene mutations that cause dyschromatosis symmetrica hereditaria result in less functional ADAR1 protein. While the function of this protein in the skin is unknown, researchers suggest that incorrect RNA editing may result in pigment-producing cells (melanocytes) that are more or less active than normal, resulting in the skin spots that occur in this disorder.

ADAR gene mutations have also been identified in individuals with various neurological problems that differ from those that occur in Aicardi-Goutières syndrome, and without the other signs and symptoms that occur in that disorder.

Other Names for This Gene

- 136 kDa double-stranded RNA-binding protein
- ADAR1
- adenosine deaminase acting on RNA 1-A
- AGS6
- DRADA
- DSH
- DSRAD
- dsRNA adenosine deaminase
- dsRNA adeonosine deaminase
- G1P1
- IFI-4
- IFI4
- interferon-induced protein 4
- interferon-inducible protein 4
- K88DSRBP
- P136

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ADAR ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=103\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=103[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ADAR%5BTIAB%5D%29+OR+%28adenosine+deaminase,+RNA+specific%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+720+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- DYSCHROMATOSIS SYMMETRICA HEREDITARIA; DSH (<https://omim.org/entry/127400>)
- ADENOSINE DEAMINASE, RNA-SPECIFIC; ADAR (<https://omim.org/entry/146920>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/103>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ADAR\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ADAR[gene]))

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Genomic Location

The *ADAR* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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