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) 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 immune system sometimes fails to recognize the body's own RNA and may act as though it belongs to a pathogen that should be attacked. By changing adenosine to inosine, ADAR1 may prevent the immune system from targeting 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 the 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

Variants (also called mutations) in the *ADAR* gene have been identified in people with Aicardi-Goutières syndrome, a disorder that typically involves severe brain dysfunction (encephalopathy), skin lesions, immune system abnormalities, and other health problems. Some of these variants lead to the production of 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

ADAR gene variants 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 the surrounding skin. These macules generally appear in infancy or early childhood.

Variants in the *ADAR* gene that lead to dyschromatosis symmetrica hereditaria cause cells to produce a version of the ADAR1 protein that does not function properly. 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, causing the skin spots that occur in people with this disorder.

Other Names for This Gene

- ADAR1
- adenosine deaminase acting on RNA 1-A
- DRADA
- DSH
- DSRAD
- dsRNA adenosine deaminase
- dsRNA adeonosine deaminase
- IFI-4
- IFI4
- interferon-induced protein 4
- interferon-inducible protein 4

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])

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])

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Genomic Location

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

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