RNASEH2A gene

ribonuclease H2 subunit A

Normal Function

The *RNASEH2A* gene provides instructions for making one part (subunit) of a group of proteins called the RNase H2 complex. This complex is a ribonuclease, which means it is an enzyme that helps break down molecules that contain RNA, a chemical cousin of DNA. In particular, the RNase H2 complex helps break down a specific type of molecule that is made up of one strand of RNA and one strand of DNA (RNA-DNA hybrids). RNA-DNA hybrids are formed during DNA copying (replication) and are found in all cells.

The RNase H2 complex is also thought to be involved in DNA replication and error repair. This complex also likely plays an important role in the immune system by removing unnecessary pieces of DNA that might otherwise trigger an immune response.

Health Conditions Related to Genetic Changes

Aicardi-Goutières syndrome

Variants (also called mutations) in the *RNASEH2A* gene have been identified in people with Aicardi-Goutières syndrome, a disorder that often involves severe brain dysfunction (encephalopathy), skin lesions, and other health problems. The *RNASEH2A* gene variants that cause Aicardi-Goutières syndrome likely produce a dysfunctional RNase H2 complex. When this complex is not functioning properly, it may disrupt transcription, DNA replication, DNA repair, cell death (apoptosis), or other processes. Such disruptions are thought to lead to the accumulation of unneeded DNA and RNA in cells. These DNA and RNA fragments may be mistaken for the genetic material of viral invaders, triggering immune system reactions in multiple body systems that cause the signs and symptoms of Aicardi-Goutières syndrome.

Other Names for This Gene

- ribonuclease H2, large subunit
- ribonuclease H2, subunit A
- ribonuclease HI large subunit
- ribonuclease HI subunit A
- RNase H(35)

- RNase H2 subunit A
- RNase HI large subunit
- RNASEHI
- RNH2A_HUMAN
- RNHIA
- RNHL

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of RNASEH2A (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10535[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28RNASEH2A%5BTIAB%5D%29+OR+%28%28AGS4%5BTIAB%5D%29+OR+%28RNHL%5BTIAB%5D%29+OR+%28RNASEHI%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)

Catalog of Genes and Diseases from OMIM

- RIBONUCLEASE H2, SUBUNIT A; RNASEH2A (https://omim.org/entry/606034)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/10535)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=RNASEH2A[gene])

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Genomic Location

The *RNASEH2A* gene is found on chromosome 19 (https://medlineplus.gov/genetics/chromosome/19/).

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