RNASEH2C gene

ribonuclease H2 subunit C

Normal Function

The *RNASEH2C* 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 containing RNA, a chemical cousin of DNA. In particular, the RNase H2 complex normally helps break down molecules in which one strand of RNA is combined with one strand of DNA (RNA-DNA hybrids) when these molecules are no longer needed. 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, error repair, and other cellular processes, including helping to prevent inappropriate immune system activation.

Health Conditions Related to Genetic Changes

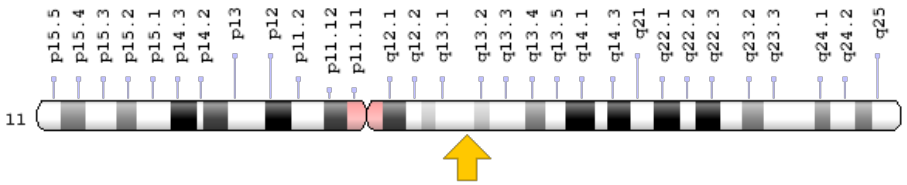
Aicardi-Goutières syndrome

At least 26 mutations in the *RNASEH2C* gene have been identified in people with Aicardi-Goutières syndrome, a disorder that involves severe brain dysfunction (encephalopathy), skin lesions, and other health problems associated with abnormal immune system activation. The *RNASEH2C* gene mutations that cause Aicardi-Goutières syndrome likely result in a dysfunctional RNase H2 complex. Abnormal functioning of this complex 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 severe brain dysfunction (encephalopathy), skin lesions, and other signs and symptoms of Aicardi-Goutières syndrome.

Chromosomal Location

Cytogenetic Location: 11q13.1, which is the long (q) arm of chromosome 11 at position 13.1

Molecular Location: base pairs 65,717,673 to 65,720,938 on chromosome 11 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AGS3
- aicardi-Goutieres syndrome 3 protein
- AYP1
- FLJ20974
- MGC22934
- ribonuclease H2, subunit C
- ribonuclease HI subunit C
- RNase H1 small subunit
- RNase H2 subunit C
- RNH2C_HUMAN

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): DNA Replication Mechanisms https://www.ncbi.nlm.nih.gov/books/NBK26850/

Clinical Information from GeneReviews

- Aicardi-Goutieres Syndrome https://www.ncbi.nlm.nih.gov/books/NBK1475

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28RNASEH2C%5BTIAB%5D%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 C
  http://omim.org/entry/610330

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RNASEH2C%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:24116
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:84153
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/84153
- UniProt
  https://www.uniprot.org/uniprot/Q8TDP1

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