ADA gene
adenosine deaminase

Normal Function

The *ADA* gene provides instructions for producing the enzyme adenosine deaminase. This enzyme is produced in all cells, but the highest levels of adenosine deaminase occur in immune system cells called lymphocytes, which develop in lymphoid tissues. These lymphoid tissues include the thymus, which is a gland located behind the breastbone, and lymph nodes, which are found throughout the body. Lymphocytes form the immune system, which defends the body against potentially harmful invaders, such as viruses or bacteria.

The function of the adenosine deaminase enzyme is to eliminate a molecule called deoxyadenosine, which is generated when DNA is broken down. Adenosine deaminase converts deoxyadenosine, which is toxic to lymphocytes, to another molecule called deoxyinosine, which is not harmful.

Health Conditions Related to Genetic Changes

Adenosine deaminase deficiency

More than 70 mutations in the *ADA* gene have been identified. Most of these mutations result in the substitution of one protein building block (amino acid) for another amino acid in the adenosine deaminase enzyme. Other mutations cause the enzyme to be unstable or prevent it from being produced at all.

These mutations result in the absence or deficiency of the adenosine deaminase enzyme in cells, preventing the normal breakdown of deoxyadenosine. A buildup of this toxic compound interferes with the development and maintenance of lymphocytes, resulting in severe combined immunodeficiency (SCID), which is characteristic of adenosine deaminase deficiency.
Chromosomal Location

Cytogenetic Location: 20q13.12, which is the long (q) arm of chromosome 20 at position 13.12

Molecular Location: base pairs 44,619,519 to 44,651,758 on chromosome 20 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

• ADA_HUMAN
• adenosine aminohydrolase

Additional Information & Resources

Clinical Information from GeneReviews

• Adenosine Deaminase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1483

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ADA%5BTIAB%5D%29+OR+%28adenosine+deaminase%5BTIAB%5D%29+OR+%28adenosine+aminohydrolase%5BMAJR%5D%29+OR+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

• ADENOSINE DEAMINASE
  http://omim.org/entry/608958
Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ADA.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ADA%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:100
- Mutation Registry for Adenosine Deaminase Deficiency (ADA)
  http://structure.bmc.lu.se/idbase/ADAbase/
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P00813

Sources for This Summary

- OMIM: ADENOSINE DEAMINASE
  http://omim.org/entry/608958
- Blackburn MR, Thompson LF. Adenosine deaminase deficiency: unanticipated benefits from
  jimmunol.1103519.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22262755
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3341658/
- Buckley RH. Molecular defects in human severe combined immunodeficiency and approaches to
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15032591
- Hershfield MS. Genotype is an important determinant of phenotype in adenosine deaminase
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14499267
- Hershfield MS. New insights into adenosine-receptor-mediated immunosuppression and the role of
  adenosine in causing the immunodeficiency associated with adenosine deaminase deficiency. Eur J
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15580654
- Nyhan WL. Disorders of purine and pyrimidine metabolism. Mol Genet Metab. 2005 Sep-Oct;
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16176880

Reprinted from Genetics Home Reference: