ACY1 gene

aminoacylase 1

Normal Function

The *ACY1* gene provides instructions for making an enzyme called aminoacylase 1, which is found in many tissues and organs, including the kidneys and the brain. This enzyme is involved in the breakdown of proteinswhen they are no longer needed. Many proteins in the body have a chemical group called an acetyl group attached to one end. This modification, called *N*-acetylation, helps protect and stabilize the protein. Aminoacylase 1 performs the final step in the breakdown of these proteins by removing the acetyl group from certain protein building blocks (amino acids). The amino acids can then be recycled and used to build other proteins.

Health Conditions Related to Genetic Changes

Aminoacylase 1 deficiency

Several mutations in the *ACY1* gene have been identified in people with a condition called aminoacylase 1 deficiency. This condition is characterized by delayed development of mental and motor skills and other neurological problems, although some people with the condition have no signs or symptoms. Most of the associated *ACY1* gene mutations change single amino acids in the aminoacylase 1 enzyme. These and other *ACY1* gene mutations lead to production of an aminoacylase 1 enzyme with little or no function. Without this enzyme's function, acetyl groups are not efficiently removed from a subset of amino acids (including methionine, glutamic acid, alanine, serine, glycine, leucine, valine, threonine, and isoleucine) during the breakdown of proteins. The excess *N*-acetylated amino acids are released from the body in urine. It is not known how a reduction of aminoacylase 1 function leads to neurological problems in people with aminoacylase 1 deficiency.

Other Names for This Gene

- ACY-1
- ACY1D
- acylase 1
- aminoacylase-1
- N-acyl-L-amino-acid amidohydrolase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ACY1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=95[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ACY1%5BTIAB%5D%29+OR+%28aminoacylase+1%5BTIAB%5D%29%29+OR+%28%28ACY-1%5BTIAB%5D%29+OR+%28aminoacylase-1%5BTIAB%5D%29+OR+%28acylase%5BTIAB%5D%29+OR+%28N-acyl-L-amino-acid+amidohydrolase%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

- AMINOACYLASE 1; ACY1 (https://omim.org/entry/104620)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/95)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ACY1[gene])

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Genomic Location

The *ACY1* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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