

AMPD1 gene

adenosine monophosphate deaminase 1

Normal Function

The *AMPD1* gene provides instructions for producing an enzyme called adenosine monophosphate (AMP) deaminase. This enzyme is found in the muscles used for movement (skeletal muscles), where it plays a role in producing energy. Specifically, during physical activity, this enzyme converts a molecule called adenosine monophosphate (AMP) to a molecule called inosine monophosphate (IMP) as part of a process called the purine nucleotide cycle. This cycle reuses molecules called purines, which are a group of building blocks of DNA (nucleotides), its chemical cousin RNA, and molecules such as AMP that serve as energy sources in the cell. As part of the purine nucleotide cycle, AMP deaminase converts AMP to IMP, and as the cycle continues, molecules are produced that the muscle cells can use for energy. Skeletal muscle cells need energy to function and move the body.

Health Conditions Related to Genetic Changes

Adenosine monophosphate deaminase deficiency

At least nine mutations in the *AMPD1* gene have been found to cause AMP deaminase deficiency. This condition is characterized by skeletal muscle pain or weakness after exercise or prolonged physical activity (exercise intolerance). Most cases are caused by a mutation that results in a premature stop signal in the instructions for making AMP deaminase (written as Gly12Ter or Q12X). The resulting enzyme is abnormally short and nonfunctional and cannot participate in the purine nucleotide cycle. As a result, the process stalls and energy production in skeletal muscle cells is decreased. Skeletal muscles are particularly sensitive to decreases in energy during periods of exercise or increased activity when energy demands increase. The lack of AMP deaminase as a source of energy production can result in fatigue and muscle weakness or pain in some people with AMP deaminase deficiency.

It is not known why some people with this condition do not experience symptoms. Researchers speculate that additional factors, both genetic and environmental, may determine whether a person develops the signs and symptoms of AMP deaminase deficiency.

Other Names for This Gene

- Adenosine monophosphate deaminase-1 (muscle)
- AMP deaminase
- AMPD1_HUMAN
- MAD
- MADA

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AMPD1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=270\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=270[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28AMPD1%5BTIAB%5D%29+OR+%28adenosine+monophosphate+deaminase+1%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+1800+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- ADENOSINE MONOPHOSPHATE DEAMINASE 1; AMPD1 (<https://omim.org/entry/102770>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/270>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=AMPD1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=AMPD1[gene]))

References

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Genomic Location

The *AMPD1* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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