

## Adenosine monophosphate deaminase deficiency

### Description

Adenosine monophosphate (AMP) deaminase deficiency is a condition that can affect the muscles used for movement (skeletal muscles). In many affected individuals, AMP deaminase deficiency does not cause any symptoms. People who do experience symptoms typically have fatigue, muscle pain (myalgia), or cramps after exercise or prolonged physical activity (exercise intolerance). Following strenuous activity, they often get tired more quickly and stay tired longer than would normally be expected. In rare cases, affected individuals have more severe symptoms including severe muscle weakness, low muscle tone (hypotonia), and muscle wasting (atrophy), but it is unclear whether these symptoms are due solely to AMP deaminase deficiency or additional health conditions. Exercise intolerance associated with AMP deaminase deficiency usually becomes apparent in childhood or early adulthood.

### Frequency

AMP deaminase deficiency is one of the most common inherited muscle disorders in white populations, affecting 1 in 50 to 100 people. The prevalence is lower in African Americans, affecting an estimated 1 in 40,000 people, and the condition is even less common in the Japanese population.

### Causes

AMP deaminase deficiency is caused by mutations in the *AMPD1* gene, which provides instructions for producing an enzyme called AMP deaminase. This enzyme is found in skeletal muscles, where it plays a role in producing energy. Skeletal muscle cells need energy to function and move the body.

Mutations in the *AMPD1* gene often result in an AMP deaminase enzyme that cannot function and as a result, 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 activity can result in fatigue, muscle weakness or pain, or other muscle problems 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.

Learn more about the gene associated with Adenosine monophosphate deaminase deficiency

- AMPD1

## **Inheritance**

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## **Other Names for This Condition**

- AMP deaminase deficiency
- Exercise-induced myopathy
- MAD deficiency
- MADA deficiency
- Muscle AMP deaminase deficiency
- Myoadenylate deaminase deficiency

## **Additional Information & Resources**

### Genetic Testing Information

- Genetic Testing Registry: Muscle AMP deaminase deficiency (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3714933/>)

### Genetic and Rare Diseases Information Center

- Adenosine monophosphate deaminase deficiency (<https://rarediseases.info.nih.gov/diseases/547/index>)

### Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

### Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Adenosine monophosphate deaminase deficiency%22](https://clinicaltrials.gov/search?cond=%22Adenosine%20monophosphate%20deaminase%20deficiency%22))

### Catalog of Genes and Diseases from OMIM

- MYOPATHY DUE TO MYOADENYLATE DEAMINASE DEFICIENCY; MMDD (<https://omim.org/entry/615511>)

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28AMP+deaminase+deficiency%5BTIAB%5D%29+OR+%28myoadenylate+deaminase+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>)

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