

## AAAS gene

aladin WD repeat nucleoporin

### Normal Function

The AAAS gene provides instructions for making a protein called ALADIN whose function is not well understood. Within cells, ALADIN is found in the nuclear envelope, the structure that surrounds the nucleus and separates it from the rest of the cell. Based on its location, ALADIN is thought to be involved in the movement of molecules into and out of the nucleus.

### Health Conditions Related to Genetic Changes

#### Triple A syndrome

At least 49 mutations in the AAAS gene have been found to cause triple A syndrome. These mutations change the structure of ALADIN in different ways; however, almost all mutations prevent this protein from reaching its proper location in the nuclear envelope. The absence of ALADIN likely disrupts the movement of molecules across this membrane. Researchers suspect that DNA repair proteins may be unable to enter the nucleus if ALADIN is missing from the nuclear envelope. DNA damage that is not repaired can cause the cell to become unstable and lead to cell death. Although the nervous system is particularly vulnerable to DNA damage, it remains unknown exactly how mutations in the AAAS gene lead to the signs and symptoms of triple A syndrome.

### Other Names for This Gene

- AAA
- AAAS\_HUMAN
- AAASb
- achalasia, adrenocortical insufficiency, alacrimia
- achalasia, adrenocortical insufficiency, alacrimia (Allgrove, triple-A)
- ADRACALA
- ADRACALIN
- ALADIN
- DKFZp586G1624

- GL003

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of AAAS ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8086\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8086[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28AAAS%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28%28aneurysm%29+OR+%28American+Association+for+the+Advancement+of+Science%29+OR+%28Time+traveling%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- ALADIN WD REPEAT NUCLEOPORIN; AAAS (<https://omim.org/entry/605378>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8086>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=AAAS\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=AAAS[gene]))

## **References**

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## Genomic Location

The AAAS gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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