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.

Chromosomal Location
Cytogenetic Location: 12q13.13, which is the long (q) arm of chromosome 12 at position 13.13

Credit: Genome Decoration Page/NCBI
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

Educational Resources

- The Cell (second edition, 2000): The Nuclear Envelope and Traffic Between the Nucleus and Cytoplasm
  https://www.ncbi.nlm.nih.gov/books/NBK9857/#A1384

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22+AND+5Bdp%5D

Catalog of Genes and Diseases from OMIM

- AAAS GENE
  http://omim.org/entry/605378

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_AAAS.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=AAAS%5Bgene%5D
- HGNC Gene Symbol Report
• Monarch Initiative
https://monarchinitiative.org/gene/NCBIGene:8086
• NCBI Gene
• UniProt
https://www.uniprot.org/uniprot/Q9NRG9

Sources for This Summary
• OMIM: AAAS GENE
http://omim.org/entry/605378

Reprinted from Genetics Home Reference:

Reviewed: February 2010
Published: June 23, 2020