ARHGAP31 gene

Rho GTPase activating protein 31

Normal Function

The *ARHGAP31* gene provides instructions for making a protein classified as a Rho GTPase activating protein (GAP). GAPs turn off (inactivate) proteins called GTPases, which play an important role in chemical signaling within cells. Often referred to as molecular switches, GTPases can be turned on and off. They are turned on (active) when they are attached (bound) to a molecule called GTP and are turned off when they are bound to another molecule called GDP. The ARHGAP31 protein inactivates GTPases known as Cdc42 and Rac1 by stimulating a reaction that turns the attached GTP into GDP. When active, Cdc42 and Rac1 transmit signals that are critical for various aspects of embryonic development. The ARHGAP31 protein appears to regulate these GTPases specifically during development of the limbs, skull, and heart.

Health Conditions Related to Genetic Changes

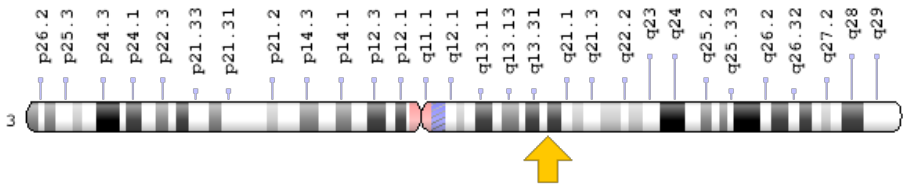
Adams-Oliver syndrome

At least three mutations in the *ARHGAP31* gene are known to cause Adams-Oliver syndrome, a condition characterized by areas of missing skin (aplasia cutis congenita), usually on the scalp, and malformations of the hands and feet. These mutations lead to production of an abnormally short ARHGAP31 protein that is more active than normal. The increased GAP activity leads to a reduction in Cdc42 and Rac1 signaling, which impairs proper development of the skin on the top of the head and the bones in the hands and feet.

Chromosomal Location

Cytogenetic Location: 3q13.32-q13.33, which is the long (q) arm of chromosome 3 between positions 13.32 and 13.33

Molecular Location: base pairs 119,294,289 to 119,420,714 on chromosome 3 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AOS1
- Cdc42 GTPase-activating protein
- CDGAP
- RHG31_HUMAN
- rho GTPase-activating protein 31

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database (2000): Rho GTPases Function as Membrane-Associated GDP/GTP-Regulated Molecular Switches https://www.ncbi.nlm.nih.gov/books/NBK6594/#_A39189_

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28ARHGAP31%5BTIAB%5D%29+OR+%28%28AOS1%5BTIAB%5D%29+OR+%28CDGAP%5BTIAB%5D%29+OR+%28Cdc42+GTPase-activating+protein%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

- RHO GTPase-ACTIVATING PROTEIN 31
  http://omim.org/entry/610911

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ARHGAP31.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ARHGAP31%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:29216
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:57514
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/57514
- UniProt
  https://www.uniprot.org/uniprot/Q2M1Z3

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Reviewed: November 2015
Published: September 10, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services