



## EOGT gene

EGF domain specific O-linked N-acetylglucosamine transferase

### Normal Function

The *EOGT* gene provides instructions for making a protein that modifies certain other proteins by transferring a molecule called N-acetylglucosamine to them. This change, called an O-GlcNAc modification, can affect protein stability and regulate several cellular processes, such as signaling in cells and the first step in the production of proteins from genes (transcription). Little is known about the proteins altered by the *EOGT* protein or what effect the O-GlcNAc modification has on them. Studies suggest that Notch proteins may be modified by *EOGT*. Notch proteins stimulate signaling pathways important during the development of several tissues throughout the body, including the bones, heart, liver, muscles, and blood cells, among others.

### Health Conditions Related to Genetic Changes

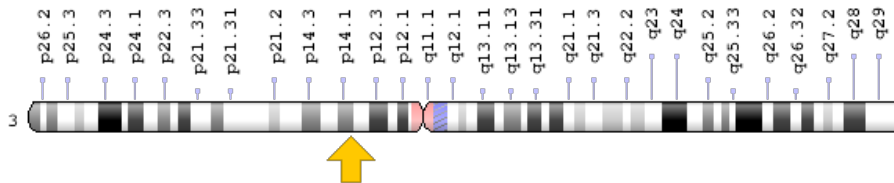
#### Adams-Oliver syndrome

At least three mutations in the *EOGT* gene have been found in individuals with Adams-Oliver syndrome. This condition is characterized by areas of missing skin (aplasia cutis congenita), usually on the scalp, and malformations of the hands and feet. The most common *EOGT* gene mutation involved in this condition, which is found in the Arab population, leads to an abnormally short protein. The other mutations change single protein building blocks (amino acids) in the *EOGT* protein. Research suggests that the *EOGT* gene mutations reduce or eliminate the protein's ability to transfer N-acetylglucosamine. It is unknown what effect this impairment has on cells or how it leads to the features of Adams-Oliver syndrome.

## Chromosomal Location

Cytogenetic Location: 3p14.1, which is the short (p) arm of chromosome 3 at position 14.1

Molecular Location: base pairs 68,975,212 to 69,013,961 on chromosome 3 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- AER61
- AER61 glycosyltransferase
- AOS4
- C3orf64
- EGF domain-specific O-linked N-acetylglucosamine (GlcNAc) transferase
- EGF domain-specific O-linked N-acetylglucosamine transferase
- EGF-O-GlcNAc transferase
- EOGT1
- EOGT\_HUMAN
- extracellular O-linked N-acetylglucosamine transferase
- FLJ33770

## Additional Information & Resources

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28EOGT%5BTIAB%5D%29+OR+%28%28C3orf64%5BTIAB%5D%29+OR+%28EOGT1%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

- EGF DOMAIN-SPECIFIC O-LINKED N-ACETYLGLUCOSAMINE TRANSFERASE  
<http://omim.org/entry/614789>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=EOGT%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report#!/hgnc\\_id/HGNC:28526](https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:28526)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:285203>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/285203>
- UniProt  
<https://www.uniprot.org/uniprot/Q5NDL2>

## **Sources for This Summary**

- Cohen I, Silberstein E, Perez Y, Landau D, Elbedour K, Langer Y, Kadir R, Volodarsky M, Sivan S, Narkis G, Birk OS. Autosomal recessive Adams-Oliver syndrome caused by homozygous mutation in EOGT, encoding an EGF domain-specific O-GlcNAc transferase. *Eur J Hum Genet.* 2014 Mar; 22(3):374-8. doi: 10.1038/ejhg.2013.159. Epub 2013 Jul 17.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/23860037>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3925282/>
- OMIM: EGF DOMAIN-SPECIFIC O-LINKED N-ACETYLGLUCOSAMINE TRANSFERASE  
<http://omim.org/entry/614789>
- Ogawa M, Sawaguchi S, Kawai T, Nadano D, Matsuda T, Yagi H, Kato K, Furukawa K, Okajima T. Impaired O-linked N-acetylglucosaminylation in the endoplasmic reticulum by mutated epidermal growth factor (EGF) domain-specific O-linked N-acetylglucosamine transferase found in Adams-Oliver syndrome. *J Biol Chem.* 2015 Jan 23;290(4):2137-49. doi: 10.1074/jbc.M114.598821. Epub 2014 Dec 8.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25488668>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4303666/>
- Sakaidani Y, Ichiyangi N, Saito C, Nomura T, Ito M, Nishio Y, Nadano D, Matsuda T, Furukawa K, Okajima T. O-linked-N-acetylglucosamine modification of mammalian Notch receptors by an atypical O-GlcNAc transferase Eogt1. *Biochem Biophys Res Commun.* 2012 Mar 2;419(1):14-9. doi: 10.1016/j.bbrc.2012.01.098. Epub 2012 Jan 28.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/22310717>

- Sakaidani Y, Nomura T, Matsuura A, Ito M, Suzuki E, Murakami K, Nadano D, Matsuda T, Furukawa K, Okajima T. O-linked-N-acetylglucosamine on extracellular protein domains mediates epithelial cell-matrix interactions. *Nat Commun.* 2011 Dec 13;2:583. doi: 10.1038/ncomms1591. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/22158438>
  - Shaheen R, Aglan M, Keppler-Noreuil K, Faqeih E, Ansari S, Horton K, Ashour A, Zaki MS, Al-Zahrani F, Cueto-González AM, Abdel-Salam G, Temtamy S, Alkuraya FS. Mutations in EOGT confirm the genetic heterogeneity of autosomal-recessive Adams-Oliver syndrome. *Am J Hum Genet.* 2013 Apr 4;92(4):598-604. doi: 10.1016/j.ajhg.2013.02.012. Epub 2013 Mar 21. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/23522784>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3617382/>
- 

Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/gene/EOGT>

Reviewed: November 2015  
Published: May 14, 2019

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