

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.

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

Tests Listed in the Genetic Testing Registry

- Tests of EOGT ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=285203\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=285203[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29>)

Catalog of Genes and Diseases from OMIM

- EGF DOMAIN-SPECIFIC O-LINKED N-ACETYLGLUCOSAMINE TRANSFERASE; EOGT (<https://omim.org/entry/614789>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/285203>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=EOGT\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=EOGT[gene]))

References

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Genomic Location

The *EOGT* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

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