

COG5 gene

component of oligomeric golgi complex 5

Normal Function

The *COG5* gene provides instructions for making a protein called component of oligomeric Golgi complex 5 (COG5). As its name suggests, COG5 is one piece of a group of proteins known as the conserved oligomeric Golgi (COG) complex. This complex functions in the Golgi apparatus, which is a cell structure in which newly produced proteins are modified. One process that occurs in the Golgi apparatus is glycosylation, by which sugar molecules (oligosaccharides) are attached to proteins and fats. Glycosylation modifies proteins so they can perform a wider variety of functions.

The COG complex takes part in the transport of proteins, including the enzymes that perform glycosylation, within the Golgi apparatus. COG is specifically involved in retrograde transport, which moves proteins backward through the Golgi apparatus. Retrograde transport is important for recycling Golgi proteins and ensuring that they are in the correct location in the structure, which is key to proper glycosylation. The proteins are transported in sac-like structures called vesicles that attach to the Golgi membrane and release the contents into the Golgi apparatus. The COG complex controls the attachment (tethering) of the vesicles to the Golgi membrane.

Health Conditions Related to Genetic Changes

COG5-congenital disorder of glycosylation

At least eight mutations in the *COG5* gene are known to cause *COG5*-congenital disorder of glycosylation (*COG5*-CDG). This condition often leads to developmental delay and intellectual disability and causes other abnormalities. Mutations in the *COG5* gene reduce the amount of COG5 protein or eliminate it completely, which disrupts retrograde transport in the Golgi apparatus. This disruption results in abnormal protein glycosylation, which can affect multiple body systems, leading to the signs and symptoms of *COG5*-CDG. The severity of the condition is related to the amount of COG5 protein that remains in cells.

Other Names for This Gene

- 13S golgi transport complex 1 90 kDa subunit
- CDG2I

- COG complex subunit 5
- conserved oligomeric Golgi complex protein 5
- conserved oligomeric Golgi complex subunit 5
- GOLTC1
- GTC90

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COG5 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10466\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10466[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28COG5%5BTIAB%5D%29+OR+%28component+of+oligomeric+golgi+complex+5%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

- COMPONENT OF OLIGOMERIC GOLGI COMPLEX 5; COG5 (<https://omim.org/entry/606821>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/10466>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=COG5\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=COG5[gene]))

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Genomic Location

The *COG5* gene is found on chromosome 7 (<https://medlineplus.gov/genetics/chromosome/7/>).

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