PLCB4 gene

phospholipase C beta 4

Normal Function

The *PLCB4* gene provides instructions for making one form (the beta 4 isoform) of a protein called phospholipase C. This protein is involved in a signaling pathway known as the phosphoinositide cycle, which helps transmit information from the outside of the cell to the inside of the cell. During this cycle, phospholipase C converts a large molecule called phosphatidylinositol 4,5-bisphosphate into two smaller molecules. These smaller molecules relay messages into the cell that ultimately influence many cell activities.

Studies suggest that the beta 4 isoform of phospholipase C contributes to the proper development of the first and second pharyngeal arches. During early development, these structures become the bones and tissues of the head and face, including the jawbones, facial muscles, and the inner and outer ear.

The beta 4 isoform of phospholipase C is also important for the proper functioning of the retina, which is the specialized light-sensitive tissue that lines the back of the eye.

Health Conditions Related to Genetic Changes

Auriculocondylar syndrome

Variants (also called mutations) in the *PLCB4* gene have been found to cause auriculocondylar syndrome. This disorder affects the development of various facial features, primarily the ears and lower jaw (mandible). Many of the *PLCB4* gene variants that cause auriculocondylar syndrome lead to the substitution of one protein building block (amino acid) for another in the beta 4 isoform of phospholipase C. These changes likely alter the structure of the protein and impair its function, leading to a disruption in cell signaling that alters the development of the structures that are formed from the first and second pharyngeal arches.

Other Names for This Gene

- ARCND2
- phospholipase C, beta 4
- PI-PLC

- PLC-beta-4

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PLCB4 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5332[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28PLCB4%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+OR+%28%28phosphoinositidase+C%5BTIAB%5D%29+AND+%28beta+4%5BTIAB%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

- PHOSPHOLIPASE C, BETA-4; PLCB4 (https://omim.org/entry/600810)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5332)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PLCB4[gene])

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Genomic Location

The *PLCB4* gene is found on chromosome 20 (https://medlineplus.gov/genetics/chromosome/20/).

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