PLCB4 Gene

Subjects: Genetics & Heredity
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phospholipase C beta 4

genes

1. Introduction

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 within cells known as the phosphoinositide cycle, which helps transmit information from outside the cell to inside the cell. Phospholipase C carries out one particular step in the phosphoinositide cycle: the conversion of a molecule called phosphatidylinositol 4,5-bisphosphate (PIP_2) to two smaller molecules, inositol 1,4,5-trisphosphate (IP_3) and 1,2-diacylglycerol. 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 development of the first and second pharyngeal arches. These embryonic structures ultimately develop into the jawbones, facial muscles, middle ear bones, ear canals, outer ears, and related tissues. This protein is also thought to play a role in vision, particularly in the function of the retina, which is a specialized tissue at the back of the eye that detects light and color.

2. Health Conditions Related to Genetic Changes

2.1. Auriculo-condylar syndrome

At least nine mutations in the *PLCB4* gene have been found to cause auriculo-condylar syndrome, a disorder that primarily affects the development of the ears and lower jaw (mandible). The identified mutations change single protein building blocks (amino acids) in the the beta 4 isoform of phospholipase C. These changes likely alter the structure of the protein and impair the phosphoinositide cycle. Abnormal signaling alters the formation of the lower jaw: instead of developing normally, the lower jaw becomes shaped more like the smaller upper jaw (maxilla). The abnormal shape leads to an unusually small chin (micrognathia) and problems with jaw function. Researchers are working to determine how mutations in this gene lead to the other developmental abnormalities associated with auriculo-condylar syndrome.

3. Other Names for This Gene

- 1-phosphatidyl-D-myo-inositol-4,5-bisphosphate
- 1-phosphatidylinositol 4,5-bisphosphate phosphodiesterase beta-4
- ARCND2
- dJ1119D9.2 (Phopholipase C, beta 4 (1-Phosphatidylinositol-4,5-Bisphosphate Phosphodiesterase Beta 4))
- inositoltrisphosphohydrolase
- monophosphatidylinositol phosphodiesterase
- phosphoinositidase C
- phosphoinositide phospholipase C-beta-4
- phospholipase C, beta 4
- PI-PLC
- PLC-beta-4
- PLCB4_HUMAN
- triphosphoinositide phosphodiesterase

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