PIK3CD gene

phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit delta

Normal Function

The *PIK3CD* gene provides instructions for making the p110 delta (p110δ) protein, which is one piece (subunit) of an enzyme called phosphatidylinositol 3-kinase (PI3K). The version of PI3K that contains the p110δ subunit, called PI3K delta, is found in white blood cells, including immune system cells (lymphocytes) called B cells and T cells. These cells recognize and attack foreign invaders, such as viruses and bacteria, to prevent infection.

PI3K delta functions as a kinase, which means that it adds a cluster of oxygen and phosphorus atoms (a phosphate group) to other proteins through a process called phosphorylation. PI3K delta phosphorylates certain signaling molecules, which triggers a series of additional reactions that transmit chemical signals within cells. In lymphocytes, PI3K delta signaling is important for many cell activities, including cell growth and division (proliferation) and maturation (differentiation). PI3K delta helps direct B cells and T cells to differentiate into different types, each of which has a distinct function in the immune system.

Health Conditions Related to Genetic Changes

Activated PI3K-delta syndrome

Variants (also called mutations) in the *PIK3CD* gene have been found to cause an immune disorder called activated PI3K-delta syndrome. *PIK3CD* gene variants cause a form of the condition called activated PI3K-delta syndrome type 1. People with activated PI3K-delta syndrome type 1 typically have recurrent bacterial infections of the respiratory tract and chronic viral infections.

The *PIK3CD* gene variants that cause activated PI3K-delta syndrome type 1 change single protein building blocks (amino acids) in the p110δ protein. These variants are classified as gain-of-function variants because a PI3K-delta enzyme that contains the altered subunit is frequently turned on (overactive). Studies indicate that this overactive signaling causes T cells to mature and die too quickly. The excess signaling also blocks maturation of B cells at an early stage; the immature B cells cannot respond to foreign invaders and likely self-destruct. The lack of T cells and B cells makes it difficult for people with this disorder to fight off bacterial and viral infections. Overactive of PI3K-

delta signaling can also stimulate the abnormal proliferation of lymphocytes, and accumulation of these cells can lead to enlarged lymph nodes (lymphadenopathy). Activated PI3K-delta syndrome type 1 is also associated with an increased risk of developing a form of blood cell cancer called lymphoma.

Autoimmune lymphoproliferative syndrome

MedlinePlus Genetics provides information about Autoimmune lymphoproliferative syndrome

Other Names for This Gene

- p110D
- P110DELTA
- phosphatidylinositol-4,5-bisphosphate 3-kinase 110 kDa catalytic subunit delta
- phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit delta
- phosphoinositide-3-kinase C
- PI3-kinase p110 subunit delta

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PIK3CD (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5293[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28PIK3CD%5BTIAB%5D%29+OR+%28%28P110DELTA%5BTIAB%5D%29+OR+%28PI3Kdelta%5BTIAB%5D%29+OR+%28P110D%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+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- PHOSPHATIDYLINOSITOL 3-KINASE, CATALYTIC, DELTA; PIK3CD (https://omim.org/entry/602839)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5293)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PIK3CD[gene])

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Genomic Location

The *PIK3CD* gene is found on chromosome 1 (https://medlineplus.gov/genetics/chromosome/1/).

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