



PIK3CD gene

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

Normal Function

The *PIK3CD* gene provides instructions for making the p110 delta (p110 δ) protein, which is one piece (subunit) of an enzyme called phosphatidylinositol 3-kinase (PI3K). The version of PI3K containing the p110 δ subunit, called PI3K-delta, is specifically 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.

Like other kinases, PI3K-delta 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

At least four mutations in the *PIK3CD* gene have been found to cause a form of immunodeficiency called activated PI3K-delta syndrome. Immunodeficiencies are conditions in which the immune system is not able to protect the body effectively from foreign invaders such as bacteria and viruses. People with activated PI3K-delta syndrome typically have recurrent bacterial infections of the respiratory tract and chronic viral infections.

The *PIK3CD* gene mutations involved in activated PI3K-delta syndrome change single protein building blocks (amino acids) in the p110 δ protein; the most common mutation replaces the amino acid glutamic acid with the amino acid lysine at position 1021 of the protein (written as Glu1021Lys or E1021K). A PI3K-delta enzyme containing the altered p110 δ subunit is abnormally turned on (activated). 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. Lack of T cells and B cells makes it difficult for people with this disorder to fight off bacterial and viral infections. Overactivation of PI3K-delta signaling can also stimulate

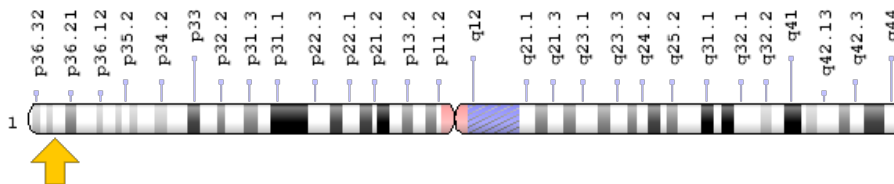
abnormal proliferation of lymphocytes, and accumulation of these cells can lead to enlarged lymph nodes (lymphadenopathy). Activated PI3K-delta syndrome also increases the risk of developing a form of cancer called B-cell lymphoma.

Autoimmune lymphoproliferative syndrome

Chromosomal Location

Cytogenetic Location: 1p36.22, which is the short (p) arm of chromosome 1 at position 36.22

Molecular Location: base pairs 9,629,889 to 9,729,114 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- APDS
- IMD14
- p110D
- P110DELTA
- phosphatidylinositol-4,5-bisphosphate 3-kinase 110 kDa catalytic subunit delta
- phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit delta isoform
- phosphatidylinositol 4,5-bisphosphate 3-kinase catalytic subunit delta isoform
- phosphatidylinositol-4,5-bisphosphate 3-kinase, catalytic subunit delta
- phosphoinositide-3-kinase C
- phosphoinositide-3-kinase, catalytic, delta polypeptide variant p37delta
- PI3-kinase p110 subunit delta
- PI3K
- PI3Kdelta
- ptdIns-3-kinase subunit p110-delta

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): PI3-Kinase Produces Inositol Phospholipid Docking Sites in the Plasma Membrane
https://www.ncbi.nlm.nih.gov/books/NBK26822/#_A2861_

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?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
<http://omim.org/entry/602839>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/PIK3CDID46261ch1p36.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PIK3CD%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:8977
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:5293>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/5293>
- UniProt
<https://www.uniprot.org/uniprot/O00329>

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