PTCH1 gene

patched 1

Normal Function

The *PTCH1* gene provides instructions for producing the patched-1 protein, which functions as a receptor. Receptor proteins have specific sites into which certain other proteins, called ligands, fit like keys into locks. A protein called Sonic Hedgehog is the ligand for the patched-1 receptor. Together, ligands and their receptors trigger signals that affect cell development and function.

Patched-1 and Sonic Hedgehog function in a pathway that is essential for early development. This pathway plays a role in cell growth, cell specialization, and determining the shape (patterning) of many different parts of the developing body. When Sonic Hedgehog is not present, patched-1 prevents cells from growing and dividing (proliferating). When Sonic Hedgehog is attached, patched-1 stops suppressing cell proliferation. Based on its role in preventing cells from proliferating in an uncontrolled way, *PTCH1* is called a tumor suppressor gene.

Health Conditions Related to Genetic Changes

Gorlin syndrome

More than 225 mutations in the *PTCH1* gene have been found to cause Gorlin syndrome (also known as nevoid basal cell carcinoma syndrome), a condition that affects many areas of the body and increases the risk of developing various cancerous and noncancerous tumors. Mutations in this gene prevent the production of patched-1 or lead to the production of an abnormal version of the receptor. An altered or missing patched-1 receptor cannot effectively suppress cell growth and division. As a result, cells proliferate uncontrollably to form the tumors that are characteristic of Gorlin syndrome. It is less clear how *PTCH1* gene mutations cause the other signs and symptoms related to this condition, including small depressions (pits) in the skin of the palms of the hands and soles of the feet, an unusually large head size (macrocephaly), and skeletal abnormalities.

9q22.3 microdeletion

The *PTCH1* gene is located in a region of chromosome 9 that is deleted in people with a 9q22.3 microdeletion. As a result of this deletion, affected individuals are missing one

copy of the *PTCH1* gene in each cell. Researchers believe that many of the features associated with 9q22.3 microdeletions, particularly the signs and symptoms of Gorlin syndrome (described above), result from a loss of the *PTCH1* gene. When this gene is missing, patched-1 is not available to suppress cell proliferation. As a result, cells divide uncontrollably to form the tumors that are characteristic of Gorlin syndrome. Other signs and symptoms related to 9q22.3 microdeletions (such as delayed development, intellectual disability, overgrowth of the body, and other physical abnormalities) may result from the loss of additional genes in the deleted region of chromosome 9.

Nonsyndromic holoprosencephaly

At least seven mutations in the *PTCH1* gene have been found to cause nonsyndromic holoprosencephaly. This condition occurs when the brain fails to divide into two halves during early development. *PTCH1* gene mutations are a rare cause of this condition. These mutations prevent the signaling that is necessary for normal brain cell patterning. The signs and symptoms of nonsyndromic holoprosencephaly are caused by abnormal development of the brain and face.

Coloboma

MedlinePlus Genetics provides information about Coloboma

Other disorders

At least seven mutations in the *PTCH1* gene have been found to cause nonsyndromic holoprosencephaly. This condition occurs when the brain fails to divide into two halves during early development. *PTCH1* gene mutations are a rare cause of nonsyndromic holoprosencephaly. These mutations prevent the signaling that is necessary for normal brain cell patterning. The signs and symptoms of nonsyndromic holoprosencephaly are caused by abnormal development of the brain and face.

Cancers

Some mutations are acquired during a person's lifetime and are present only in certain cells. These genetic changes, called somatic mutations, are not inherited. Somatic mutations in both copies of the *PTCH1* gene are associated with a non-hereditary (sporadic) type of skin cancer called basal cell carcinoma. Other sporadic types of cancer may be associated with somatic mutations in the *PTCH1* gene, including some forms of skin cancer, a childhood brain tumor called medulloblastoma, breast cancer, and colon cancer. A noncancerous (benign) jaw tumor called a keratocystic odontogenic tumor can also be associated with somatic *PTCH1* gene mutations.

Other Names for This Gene

- BCNS
- FLJ26746
- FLJ42602

- HPE7
- NBCCS
- patched
- patched homolog 1 (Drosophila)
- PTC
- PTC1
- PTC1_HUMAN
- PTCH

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PTCH1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5727[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PTCH1%5BTIAB%5D%29+OR+%28patched1%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- PATCHED 1; PTCH1 (https://omim.org/entry/601309)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5727)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PTCH1[gene])

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Genomic Location

The *PTCH1* gene is found on chromosome 9 (https://medlineplus.gov/genetics/chromosome/9/).

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