# AKT1 gene

AKT serine/threonine kinase 1

## Normal Function

The *AKT1* gene provides instructions for making a protein called AKT1 kinase. This protein is found in various cell types throughout the body, where it plays a critical role in many signaling pathways. For example, AKT1 kinase helps regulate cell growth and division (proliferation), the process by which cells mature to carry out specific functions ( differentiation), and cell survival. AKT1 kinase also helps control apoptosis, which is the self-destruction of cells when they become damaged or are no longer needed.

Signaling involving AKT1 kinase appears to be essential for the normal development and function of the nervous system. Studies have suggested a role for AKT1 kinase in cell-to-cell communication among nerve cells (neurons), neuronal survival, and the formation of memories.

The *AKT1* gene belongs to a class of genes known as oncogenes. When mutated, oncogenes have the potential to cause normal cells to become cancerous.

## Health Conditions Related to Genetic Changes

Proteus syndrome

At least one mutation in the *AKT1* gene has been found to cause Proteus syndrome, a rare condition characterized by overgrowth of the bones, skin, and other tissues. This mutation changes a single protein building block (amino acid) in AKT1 kinase. Specifically, it replaces the amino acid glutamic acid with the amino acid lysine at protein position 17 (written as Glu17Lys or E17K). The mutation is not inherited from a parent; in people with Proteus syndrome, the mutation arises randomly in one cell during the early stages of development before birth. As cells continue to grow and divide, some cells will have the mutation and other cells will not. This mixture of cells with and without a genetic mutation is known as mosaicism.

The Glu17Lys mutation leads to the production of an overactive AKT1 kinase that is turned on when it should not be. The abnormally active protein disrupts a cell's ability to regulate its own growth, allowing the cell to grow and divide abnormally. Increased cell proliferation in various tissues and organs leads to the overgrowth characteristic of Proteus syndrome. Studies suggest that the *AKT1* gene mutation is more common in

groups of cells that experience overgrowth than in the parts of the body that grow normally.

Cowden syndrome

MedlinePlus Genetics provides information about Cowden syndrome

Ovarian cancer

MedlinePlus Genetics provides information about Ovarian cancer

Schizophrenia

MedlinePlus Genetics provides information about Schizophrenia

Cancers

The Glu17Lys mutation in the *AKT1* gene (described above) has also been found in a small percentage of breast, ovarian, and colorectal cancers. In these cases the mutation is somatic, which means it is acquired during a person's lifetime and is present only in tumor cells. The mutation abnormally activates AKT1 kinase, allowing cells to grow and divide without control or order. This disordered cell proliferation leads to the development of cancerous tumors.

Although the Glu17Lys mutation has been reported in only a few types of cancer, increased activity (expression) of the *AKT1* gene is found in many types of cancer.

Other disorders

Several common variations (polymorphisms) in the *AKT1* gene have been found more often in people with schizophrenia than in those without the disease. These polymorphisms alter single DNA building blocks (nucleotides) in the *AKT1* gene. It is unknown whether the genetic changes have an effect on the structure or function of AKT1 kinase, and if so, how they are related to the development of schizophrenia. *AKT1* gene polymorphisms appear to be one of many genetic and environmental factors that contribute to the development of this complex psychiatric disorder.

## Other Names for This Gene

- AKT
- AKT1_HUMAN
- MGC99656
- PKB
- PKB alpha
- PKB-ALPHA
- PRKBA
- protein kinase B alpha

- proto-oncogene c-Akt
- RAC
- rac protein kinase alpha
- RAC-ALPHA
- RAC-alpha serine/threonine-protein kinase
- RAC-PK-alpha
- v-akt murine thymoma viral oncogene homolog 1

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AKT1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=207[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28AKT1%5BTI%5D%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

- BREAST CANCER (https://omim.org/entry/114480)
- COLORECTAL CANCER; CRC (https://omim.org/entry/114500)
- AKT SERINE/THREONINE KINASE 1; AKT1 (https://omim.org/entry/164730)
- OVARIAN CANCER (https://omim.org/entry/167000)
- SCHIZOPHRENIA; SCZD (https://omim.org/entry/181500)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/207)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AKT1[gene])

## References


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## Genomic Location

The *AKT1* gene is found on chromosome 14 (https://medlineplus.gov/genetics/chromosome/14/).

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