# MYCN gene

MYCN proto-oncogene, bHLH transcription factor

## Normal Function

The *MYCN* gene provides instructions for making a protein that plays an important role in the formation of tissues and organs during development before birth. Studies in animals suggest that this protein is necessary for normal development of the limbs, heart, kidneys, lungs, nervous system, and digestive system. The MYCN protein regulates the activity of other genes by attaching (binding) to specific regions of DNA and controlling the first step of protein production (transcription). On the basis of this action, this protein is called a transcription factor.

The *MYCN* gene belongs to a class of genes known as oncogenes. When mutated, oncogenes have the potential to cause normal cells to become cancerous. The *MYCN* gene is a member of the Myc family of oncogenes. These genes play important roles in regulating cell growth and division (proliferation) and the self-destruction of cells ( apoptosis).

## Health Conditions Related to Genetic Changes

Feingold syndrome

At least 36 mutations involving the *MYCN* gene have been found to cause Feingold syndrome type 1. This developmental disorder is characterized by abnormalities of the fingers and toes, particularly shortening of the second and fifth fingers ( brachymesophalangy). Other common features include a blockage in part of the digestive system (gastrointestinal atresia), an unusually small head size (microcephaly) and learning disabilities. Most of these mutations lead to a premature stop signal in the instructions for making the protein. In some cases of Feingold syndrome type 1, the entire *MYCN* gene is deleted. These genetic changes prevent one copy of the gene in each cell from producing any functional MYCN protein. As a result, only half the normal amount of this protein is available to control the activity of specific genes during development. It is unclear how a reduced amount of the MYCN protein causes the varied features of Feingold syndrome type 1.

Neuroblastoma

Some gene mutations are acquired during a person's lifetime and are present only in

certain cells. These changes, which are not inherited, are called somatic mutations. Somatic mutations sometimes occur when DNA makes a copy of itself (replicates) in preparation for cell division. Errors in the replication process can result in one or more extra copies of a gene within a cell. The presence of extra copies of certain genes, known as gene amplification, can underlie the formation and growth of tumor cells. For example, amplification of the *MYCN* gene is found in about 25 percent of neuroblastomas. Neuroblastoma is a type of cancerous tumor that arises in developing nerve cells. The number of copies of the *MYCN* gene varies widely among these tumors but is typically between 50 and 100. Amplification of the *MYCN* gene is associated with a severe form of neuroblastoma. It is unknown how amplification of this gene contributes to the aggressiveness of neuroblastoma.

Retinoblastoma

MedlinePlus Genetics provides information about Retinoblastoma

## Other Names for This Gene

- bHLHe37
- MYCN_HUMAN
- MYCNOT
- N-myc
- N-myc proto-oncogene protein
- neuroblastoma MYC oncogene
- neuroblastoma-derived v-myc avian myelocytomatosis viral related oncogene
- NMYC
- oncogene NMYC
- pp65/67
- v-myc avian myelocytomatosis viral oncogene neuroblastoma derived homolog
- v-myc avian myelocytomatosis viral related oncogene, neuroblastoma derived
- v-myc myelocytomatosis viral related oncogene, neuroblastoma derived
- v-myc myelocytomatosis viral related oncogene, neuroblastoma derived (avian)

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MYCN (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4613[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28MYCN%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

- MYCN PROTOONCOGENE, bHLH TRANSCRIPTION FACTOR; MYCN (https://omim.org/entry/164840)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4613)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MYCN[gene])

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## Genomic Location

The *MYCN* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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