

MUTYH gene

mutY DNA glycosylase

Normal Function

The *MUTYH* gene provides instructions for making an enzyme called MYH glycosylase, which is involved in the repair of DNA. This enzyme corrects particular errors that are made when DNA is copied (DNA replication) in preparation for cell division. DNA is made up of building blocks called nucleotides, each of which has a specific partner. Normally, adenine pairs with thymine (written as A-T) and guanine pairs with cytosine (written as G-C). During normal cellular activities, guanine sometimes becomes altered by oxygen, which causes it to pair with adenine instead of cytosine. MYH glycosylase fixes this error so mutations do not accumulate in the DNA and lead to tumor formation. This type of repair is known as base excision repair.

Health Conditions Related to Genetic Changes

Familial adenomatous polyposis

Mutations in the *MUTYH* gene cause an autosomal recessive form of familial adenomatous polyposis (also called MUTYH-associated polyposis). Mutations in this gene affect the ability of cells to correct errors made during DNA replication. In individuals who have autosomal recessive familial adenomatous polyposis, both copies of the *MUTYH* gene in each cell are mutated. Most mutations in this gene result in the production of a nonfunctional or low-functioning MYH glycosylase. When base excision repair in the cell is impaired, mutations in other genes build up, leading to cell overgrowth and possibly tumor formation. Two mutations that change the sequence of the building blocks of proteins (amino acids) in MYH glycosylase are common in people of European descent. One mutation replaces the amino acid tyrosine with the amino acid cysteine at position 179 (written as Tyr179Cys or Y179C). The other mutation switches the amino acid glycine with the amino acid aspartic acid at position 396 (written as Gly396Asp or G396D).

Other Names for This Gene

- hMYH
- mutY (E. coli) homolog
- mutY homolog

- mutY homolog (E. coli)
- MUTYH_HUMAN
- MYH

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MUTYH ([https://www.ncbi.nlm.nih.gov/qtr/all/tests/?term=4595\[geneid\]](https://www.ncbi.nlm.nih.gov/qtr/all/tests/?term=4595[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MUTYH%5BTIAB%5D%29+OR+%28mutY+homolog%5BTIAB%5D%29%29+OR+%28%28mutY+homolog%5BTIAB%5D%29+OR+%28MYH%5BTIAB%5D%29+OR+%28mutY+++homolog%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

- MutY DNA GLYCOSYLASE; MUTYH (<https://omim.org/entry/604933>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4595>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MUTYH\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MUTYH[gene]))

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Genomic Location

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

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