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 MYH-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).
Chromosomal Location

Cytogenetic Location: 1p34.1, which is the short (p) arm of chromosome 1 at position 34.1

Molecular Location: base pairs 45,329,242 to 45,340,470 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Other Names for This Gene

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

Additional Information & Resources

Educational Resources
- Genomes (second edition, 2002): Base excision repairs many types of damaged nucleotide
  https://www.ncbi.nlm.nih.gov/books/NBK21114/#A8439

Clinical Information from GeneReviews
- MUTYH-Associated Polyposis
  https://www.ncbi.nlm.nih.gov/books/NBK107219
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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+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, E. COLI, HOMOLOG OF
  http://omim.org/entry/604933

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/MUTYHID41464ch1p34.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MUTYH%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:4595
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q9UIF7

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12915454
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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1226182/
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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14633673

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15290654

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16287072

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12853198

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16042573

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12606733
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15188161

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Reprinted from Genetics Home Reference:

Reviewed: April 2008
Published: July 16, 2019

Lister Hill National Center for Biomedical Communications
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