TYRP1 gene

tyrosinase related protein 1

Normal Function

The TYRP1 gene provides instructions for making an enzyme called tyrosinase-related protein 1. This enzyme is located in melanocytes, which are specialized cells that produce a pigment called melanin. Melanin is the substance that gives skin, hair, and eyes their color. Melanin is also found in the light-sensitive tissue at the back of the eye (the retina), where it plays a role in normal vision.

Tyrosinase-related protein 1 is involved in the production of melanin, although its exact functions are unclear. Studies suggest that this enzyme may help stabilize tyrosinase, which is the enzyme responsible for the first step in melanin production. Tyrosinase-related protein 1 may also help determine the shape of melanosomes, which are the structures in melanocytes where melanin is produced.

Health Conditions Related to Genetic Changes

Oculocutaneous albinism

A small number of mutations in the TYRP1 gene have been found to cause oculocutaneous albinism type 3. This condition includes a form of albinism called rufous oculocutaneous albinism, which has been described primarily in dark-skinned people from southern Africa. Affected individuals have reddish-brown skin, ginger or red hair, and hazel or brown irises. Two TYRP1 mutations are known to cause this form of albinism in individuals from Africa. One mutation replaces a protein building block (amino acid) in tyrosine-related protein 1 with a signal that prematurely stops protein production. This mutation, written as Ser166Ter or S166X, affects the amino acid serine at protein position 166. The other mutation, written as 368delA, deletes a single DNA building block from the TYRP1 gene. Other alterations in this gene have been reported in a few affected people of non-African heritage. Most TYRP1 mutations lead to the production of an abnormally short, nonfunctional version of tyrosinase-related protein 1. Because this enzyme plays a role in normal pigmentation, its loss leads to the changes in skin, hair, and eye coloration that are characteristic of oculocutaneous albinism.

Melanoma
Chromosomal Location

Cytogenetic Location: 9p23, which is the short (p) arm of chromosome 9 at position 23

Molecular Location: base pairs 12,693,375 to 12,710,285 on chromosome 9 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- b-PROTEIN
- CAS2
- Catalase B
- CATB
- DHICA oxidase
- Glycoprotein 75
- GP75
- TRP
- TRP-1
- tyrosinase-related protein 1
- TYRP
- TYRP1_HUMAN

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TYRP1%5BTIAB%5D %29+OR+%28tyrosinase-related+protein+1%5BTIAB%5D%29+OR+%28b-PROTEIN%5BTIAB%5D%29+OR+%28TYRP%5BTIAB%5D%29+OR+%28DHICA+oxidase%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D %29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla %5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D
Catalog of Genes and Diseases from OMIM

- TYROSINASE-RELATED PROTEIN 1
  http://omim.org/entry/115501

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/TYRP1ID46370ch9p23.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=TYRP1%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:7306
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P17643

Sources for This Summary

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