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

MedlinePlus Genetics provides information about Melanoma

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

Tests Listed in the Genetic Testing Registry

- Tests of TYRP1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7306[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TYRP1%5BTIAB%5D%29+OR+%28tyrosinase-related+protein+1%5BTIAB%5D%29%29+OR+%28%28b-PROTEIN%5BTIAB%5D%29+OR+%28TYRP%5BTIAB%5D%29+OR+%28DHICA+oxidase%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TYROSINASE-RELATED PROTEIN 1; TYRP1 (https://omim.org/entry/115501)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/7306)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TYRP1[gene])

References

- Box NF, Wyeth JR, Mayne CJ, O'Gorman LE, Martin NG, Sturm RA. Completesequence and polymorphism study of the human TYRP1 gene encodingtyrosinase-related protein 1. Mamm Genome. 1998 Jan;9(1):50-3. doi:10.1007/s003359900678. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9434945)
- Forshew T, Khaliq S, Tee L, Smith U, Johnson CA, Mehdi SQ, Maher ER. Identification of novel TYR and TYRP1 mutations in oculocutaneous albinism. ClinGenet. 2005 Aug;68(2):182-4. doi: 10.1111/j.1399-0004.2005.00460.x. No abstractavailable. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15996218)
- Manga P, Kromberg JG, Box NF, Sturm RA, Jenkins T, Ramsay M. Rufousoculocutaneous albinism in southern African Blacks is caused by mutations in theTYRP1 gene. Am J Hum Genet. 1997 Nov;61(5):1095-101. doi: 10.1086/301603. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/9345097) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1716031/)
- Murisier F, Beermann F. Genetics of pigment cells: lessons from the tyrosinasegene family. Histol Histopathol. 2006 May;21(5):567-78. doi: 10.14670/HH-21.567. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16493586)
- Rooryck C, Roudaut C, Robine E, Musebeck J, Arveiler B. Oculocutaneousalbinism with TYRP1 gene mutations in a Caucasian patient. Pigment Cell Res. 2006Jun;19(3):239-42. doi: 10.1111/j.1600-0749.2006.00298.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16704458)
- Sarangarajan R, Boissy RE. Tyrp1 and oculocutaneous albinism type 3. PigmentCell Res. 2001 Dec;14(6):437-44. doi: 10.1034/j.1600-0749.2001.140603.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/11775055)

Genomic Location

The *TYRP1* gene is found on chromosome 9 (https://medlineplus.gov/genetics/chromosome/9/).

Last updated March 1, 2007