TYR gene

tyrosinase

Normal Function

The *TYR* gene provides instructions for making an enzyme called tyrosinase. 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 is responsible for the first step in melanin production. It converts a protein building block (amino acid) called tyrosine to another compound called dopaquinone. A series of additional chemical reactions convert dopaquinone to melanin in the skin, hair follicles, the colored part of the eye (the iris), and the retina.

Health Conditions Related to Genetic Changes

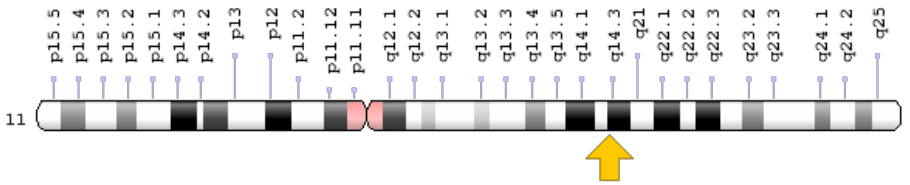
oculocutaneous albinism

More than 100 mutations in the *TYR* gene have been identified in people with oculocutaneous albinism type 1. These mutations disrupt the normal production of melanin, which reduces coloring of the hair, skin, and eyes and causes problems with vision. Most *TYR* mutations eliminate the activity of tyrosinase, preventing melanocytes from producing any melanin throughout life. These mutations cause a form of oculocutaneous albinism called type 1A (OCA1A). People with this form of albinism have white hair, light-colored eyes, and very pale skin that does not tan. Other mutations in the *TYR* gene reduce but do not eliminate tyrosinase activity. These mutations, which allow some melanin to be produced, cause oculocutaneous albinism type 1B (OCA1B). People with type 1B are also born with white hair, light-colored eyes, and pale skin, but hair and eye color often darken over time and skin may tan.

Chromosomal Location

Cytogenetic Location: 11q14.3, which is the long (q) arm of chromosome 11 at position 14.3

Molecular Location: base pairs 89,177,565 to 89,295,759 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- LB24-AB
- Monophenol monooxygenase
- OCA1A
- OCAIA
- SK29-AB
- Tumor Rejection Antigen AB
- TYRO_HUMAN

Additional Information & Resources

GeneReviews

- Oculocutaneous Albinism Type 1
  https://www.ncbi.nlm.nih.gov/books/NBK1166

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TYR%5BTI%5D%29+OR+%28tyrosinase%5BTI%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

OMIM

- TYROSINASE
  http://omim.org/entry/606933

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/TYRID42738ch11q14.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=TYR%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12442
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7299
- UniProt
  http://www.uniprot.org/uniprot/P14679

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12753405
- Oetting WS. The tyrosinase gene and oculocutaneous albinism type 1 (OCA1): A model for understanding the molecular biology of melanin formation. Pigment Cell Res. 2000 Oct;13(5):320-5. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11041207
- Opitz S, Käsmann-Kellner B, Kaufmann M, Schwinger E, Zühlke C. Detection of 53 novel DNA variations within the tyrosinase gene and accumulation of mutations in 17 patients with albinism. Hum Mutat. 2004 Jun;23(6):630-1.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15146472

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