



GPR143 gene

G protein-coupled receptor 143

Normal Function

The *GPR143* gene, also known as *OA1*, provides instructions for making a protein that is involved in the coloring (pigmentation) of the eyes and skin. This protein is made in the light-sensitive tissue at the back of the eye (the retina) and in skin cells. The GPR143 protein is part of a signaling pathway that controls the growth and maturation of melanosomes, which are cellular structures that produce and store a pigment called melanin. Melanin is the substance that gives skin, hair, and eyes their color. In the retina, this pigment also plays a critical role in normal vision.

Health Conditions Related to Genetic Changes

Ocular albinism

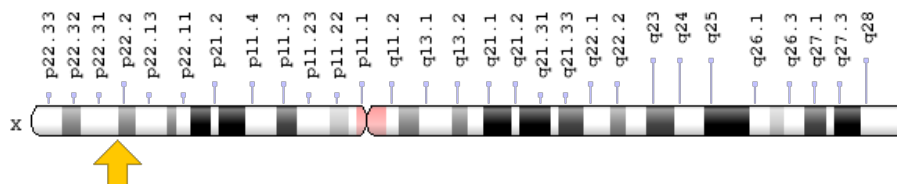
More than 60 *GPR143* mutations have been identified in people with the most common form of ocular albinism, which is called the Nettleship-Falls type or type 1. Most mutations alter the size or shape of the GPR143 protein. These genetic changes often prevent the abnormal protein from ever reaching melanosomes, where it is needed to control the growth of these pigment-containing structures. In other cases, the GPR143 protein reaches melanosomes normally but mutations prevent the protein from interacting with other molecules in its signaling pathway. Without functional GPR143 protein, melanosomes in skin cells and the retina can grow abnormally large. It is unclear how these giant melanosomes (macromelanosomes) are related to vision loss and other eye abnormalities in people with ocular albinism.

Most forms of albinism result from a reduced amount of melanin pigment within cells. Researchers continue to study why ocular albinism occurs when cells in the retina appear to contain a substantial amount of melanin. It is possible that this pigment is concentrated into a few abnormal macromelanosomes instead of being evenly distributed among many normal-sized melanosomes within the cell. Additional studies may help clarify the relationship between melanosomes, melanin distribution, and the reduced levels of pigmentation that are characteristic of ocular albinism.

Chromosomal Location

Cytogenetic Location: Xp22.2, which is the short (p) arm of the X chromosome at position 22.2

Molecular Location: base pairs 9,725,346 to 9,786,260 on the X chromosome (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- GP143_HUMAN
- OA1
- ocular albinism 1 (Nettleship-Falls)
- ocular albinism type 1 protein

Additional Information & Resources

Clinical Information from GeneReviews

- Ocular Albinism, X-Linked
<https://www.ncbi.nlm.nih.gov/books/NBK1343>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28GPR143%5BTIAB%5D%29+OR+%28%28OA1%5BTIAB%5D%29+OR+%28ocular+albinism+1%5BTIAB%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- G PROTEIN-COUPLED RECEPTOR 143
<http://omim.org/entry/300808>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_GPR143.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=GPR143%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:20145
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:4935>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4935>
- UniProt
<https://www.uniprot.org/uniprot/P51810>

Sources for This Summary

- Camand O, Boutboul S, Arbogast L, Roche O, Sternberg C, Sutherland J, Levin A, Héon E, Menasche M, Dufier J, Abitbol M. Mutational analysis of the OA1 gene in ocular albinism. *Ophthalmic Genet.* 2003 Sep;24(3):167-73.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12868035>
- Cortese K, Giordano F, Surace EM, Venturi C, Ballabio A, Tacchetti C, Marigo V. The ocular albinism type 1 (OA1) gene controls melanosome maturation and size. *Invest Ophthalmol Vis Sci.* 2005 Dec;46(12):4358-64.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16303920>
- Innamorati G, Piccirillo R, Bagnato P, Palmisano I, Schiaffino MV. The melanosomal/lysosomal protein OA1 has properties of a G protein-coupled receptor. *Pigment Cell Res.* 2006 Apr;19(2):125-35.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16524428>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1459912/>
- Lewis RA. Ocular Albinism, X-Linked. 2004 Mar 12 [updated 2015 Nov 19]. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Ledbetter N, Mefford HC, Smith RJH, Stephens K, editors. *GeneReviews®* [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1343/>
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20301517>
- Mayeur H, Roche O, Vêtu C, Jaliffa C, Marchant D, Dollfus H, Bonneau D, Munier FL, Schorderet DF, Levin AV, Héon E, Sutherland J, Lacombe D, Said E, Mezer E, Kaplan J, Dufier JL, Marsac C, Menasche M, Abitbol M. Eight previously unidentified mutations found in the OA1 ocular albinism gene. *BMC Med Genet.* 2006 Apr 28;7:41.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16646960>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1468396/>
- Oetting WS. New insights into ocular albinism type 1 (OA1): Mutations and polymorphisms of the OA1 gene. *Hum Mutat.* 2002 Feb;19(2):85-92. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11793467>

- Schiaffino MV, Tacchetti C. The ocular albinism type 1 (OA1) protein and the evidence for an intracellular signal transduction system involved in melanosome biogenesis. *Pigment Cell Res.* 2005 Aug;18(4):227-33. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16029416>
 - Shen B, Samaraweera P, Rosenberg B, Orlow SJ. Ocular albinism type 1: more than meets the eye. *Pigment Cell Res.* 2001 Aug;14(4):243-8. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11549106>
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