## Open Peer Review on Qeios

## **MITF wt Allele**

National Cancer Institute

## Source

National Cancer Institute. <u>MITF wt Allele</u>. NCI Thesaurus. Code C70585.

Human MITF wild-type allele is located within 3p14.2-p14.1 and is approximately 229 kb in length. This allele, which encodes microphthalmia-associated transcription factor protein, plays a role in both the differentiation of various cell types and the development of melanocytes in the retinal pigment epithelium. Deletion mutations in this gene are associated with both Waardenburg syndrome type IIA and Tietz syndrome. Elevated expression of this gene has been found in chemotherapy-resistant melanomas.