

Product name:	MITF Rabbit Polyclonal Antibody
Cat number:	ABN13918
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human MITF. AA range:151-200
Reactivity:	Human,Mouse
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:200-1:1000,ELISA 1:5000-1:20000
Molecular Weight:	52kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

This gene encodes a transcription factor that contains both basic helix-loop-helix and leucine zipper structural features. It regulates the differentiation and development of melanocytes retinal pigment epithelium and is also responsible for pigment cell-specific transcription of the melanogenesis enzyme genes. Heterozygous mutations in the this gene cause auditory-pigmentary syndromes, such as Waardenburg syndrome type 2 and Tietz syndrome. Alternatively spliced transcript variants encoding different isoforms have been identified. [provided by RefSeq, Jul 2008],alternative products:The X2-type isoforms differ from the X1-type isoforms by the absence of a 6 residue insert,disease:Defects in MITF are a cause of Waardenburg syndrome type 2 with ocular albinism (WS2-OA) [MIM:103470]. It is an ocular albinism with sensorineural deafness.,disease:Defects in MITF are the cause of Tietz syndrome [MIM:103500]. It is an autosomal dominant disorder characterized by generalized hypopigmentation and profound, congenital, bilateral deafness. Penetrance is complete.,disease:Defects in MITF are the cause of Waardenburg syndrome type 2A (WS2A) [MIM:193510]. It is a dominant inherited disorder characterized by sensorineural hearing loss and patches of depigmentation. The features show variable expression and penetrance.,function:Transcription factor for tyrosinase and tyrosinase-related protein 1. Binds to a symmetrical DNA sequence (E-boxes) (5'-CACGTG-3') found in the tyrosinase promoter. Plays a critical role in the differentiation of various cell types as neural crest-derived melanocytes, mast cells, osteoclasts and optic cup-derived retinal pigment epithelium.,PTM:Phosphorylation at Ser-405 significantly enhances the ability to bind the tyrosinase promoter.,similarity:Belongs to the Mit/TFE family.,similarity:Contains 1 basic helix-loop-helix (bHLH) domain.,subunit:Efficient DNA binding requires dimerization with another bHLH protein. Binds DNA in the form of homodimer or heterodimer with either TFE3, TFEB or TFEC.,tissue specificity:Isoform M is exclusively expressed in melanocytes and melanoma cells. Isoform A and isoform H are widely expressed in many cell types including melanocytes and retinal pigment epithelium (RPE). Isoform C is expressed in many cell types including RPE but not in melanocyte-lineage cells.,