10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

MITF Rabbit pAb

Catalog # AP100199

Product Information

Application	WB
Primary Accession	O75030
Other Accession	O75030
Reactivity	Human, Mouse
Predicted	Rat, Rabbit, Chicken, Dog, Horse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	58795
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human MITF
Epitope Specificity	351-450/526
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the MiT/TFE family.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. Interacts with KARS.
Post-translational modifications	Phosphorylation at Ser-405 significantly enhances the ability to bind the tyrosinase promoter. Phosphorylated at Ser-180 and Ser-516 following KIT signaling, trigerring a short live activation: Phosphorylation at Ser-180 and Ser-516 by MAPK and RPS6KA1, respectively, activate the transcription factor activity but also promote ubiquitination and subsequent degradation by the proteasome. Ubiquitinated following phosphorylation at Ser-180, leading to subsequent degradation by the proteasome. Deubiquitinated by USP13, preventing its degradation.
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. [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 (TIETZS) [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 a cause of susceptibility to cutaneous malignant melanoma type 8 (CMM8) [MIM:614456]. A malignant neoplasm of melanocytes, arising de novo or from a pre-existing benign nevus, which occurs most often in the skin but also may involve other sites.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	The protein encoded by this gene is a transcription factor that contains both basic helix-loop-helix and leucine zipper structural features. The encoded protein regulates melanocyte development and is 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. [provided by RefSeq, Aug 2017]

Additional Information

Gene ID	4286
Other Names	CMM8; COMMAD; MI; MITF-A; WS2; WS2A; bHLHe32; BCC2; Gsfbcc2; Vitiligo; Wh; bw; vit; MITF_HUMAN; MITF; Class E basic helix-loop-helix protein 32 (bHLHe32); MITF_MOUSE; MITF_RAT; melanocyte inducing transcription factor; Waardenburg syndrome, type 2A; microphthalmia-associated transcription factor; melanogenesis associated transcription factor; homolog of mouse microphthalmia
Dilution	WB=1:500-2000,ICC/IF=1:50-200
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	MITF {ECO:0000303|PubMed:8069297, ECO:0000312|HGNC:HGNC:7105}
Function	Transcription factor that acts as a master regulator of melanocyte survival and differentiation as well as melanosome biogenesis (PubMed:10587587, PubMed:22647378, PubMed:27889061, PubMed:9647758). Binds to M-boxes (5'-TCATGTG-3') and symmetrical DNA sequences (E-boxes) (5'-CACGTG-3') found in the promoter of pigmentation genes, such as tyrosinase (TYR) (PubMed:10587587, PubMed:22647378, PubMed:27889061, PubMed:9647758). Involved in the cellular response to amino acid availability by acting downstream of MTOR: in the presence of nutrients, MITF phosphorylation by MTOR promotes its inactivation (PubMed:36608670). Upon starvation or lysosomal stress, inhibition of MTOR induces MITF dephosphorylation, resulting in transcription factor activity (PubMed:36608670). Plays an important role in melanocyte development by regulating the expression of tyrosinase (TYR) and tyrosinase-related protein 1 (TYRP1) (PubMed:10587587, PubMed:22647378, PubMed:27889061, PubMed:9647758). Plays a critical role in the differentiation of various cell types, such as neural crest-derived melanocytes, mast cells, osteoclasts and optic cup-derived retinal pigment epithelium (PubMed:10587587, PubMed:22647378, PubMed:27889061, PubMed:9647758).
Cellular Location	Nucleus. Cytoplasm. Lysosome membrane Note=When nutrients are present, recruited to the lysosomal membrane via association with GDP-bound RagC/RRAGC (or RagD/RRAGD): it is then phosphorylated by MTOR (PubMed:23401004, PubMed:36608670) Phosphorylation by MTOR promotes ubiquitination and degradation (PubMed:36608670). Conversely, inhibition of mTORC1, starvation and lysosomal disruption, promotes dephosphorylation and translocation to the nucleus (PubMed:36608670). Phosphorylation by MARK3/cTAK1 promotes association with 14-3-3/YWHA adapters and retention in the cytosol (PubMed:16822840).
Tissue Location	Expressed in melanocytes (at protein level). [Isoform C2]: Expressed in the

kidney and retinal pigment epithelium. [Isoform H2]: Expressed in the kidney. [Isoform Mdel]: Expressed in melanocytes.

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