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Rabbit Anti-MITF antibody

SL1990R

Product Name:	MITF
Chinese Name:	MITF相关转录因子抗体
Alias:	Class E basic helix-loop-helix protein 32; BHLHE32; bHLHe32; Class E basic helix-loop-helix protein 32; CMM8; Homolog of mouse microphthalmia; Mi; Microphthalmia associated transcription factor; Microphthalmia, mouse, homolog of; Microphthalmia-associated transcription factor; MITF_HUMAN; WS2; WS2A.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Cow,Horse,Rabbit,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	59kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human MITF:351-450/526
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. 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.
PubMed:	PubMed
Product Detail:	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].

Function:
Transcription factor for tyrosinase (TYR) and tyrosinase-related protein 1 (TYRP1) that plays a key role in melanocyte development. 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.

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.

Subcellular Location:
Nucleus.

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.

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.

Similarity:
Belongs to the MiT/TFE family.
Contains 1 basic helix-loop-helix (bHLH) domain.

SWISS:
O75030

Gene ID:
4286

Database links:

Entrez Gene: 4286Human

Entrez Gene: 17342Mouse

Entrez Gene: 25094Rat

Omim: 156845Human

SwissProt: O75030Human

SwissProt: Q08874Mouse

SwissProt: O88368Rat

Unigene: 166017Human

Unigene: 618266Human

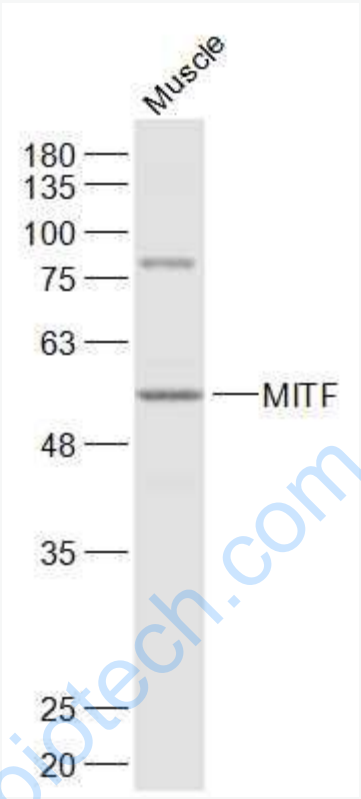
Unigene: 333284Mouse

Unigene: 454504Mouse

Unigene: 31427Rat

Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

MITF微小转录因子是一个黑色素细胞的核蛋白,对黑色素细胞的生成和活性起着关键作用,MITF也是控制细胞外信号的一项调节因子。MITF高度表达于原发和转移的恶性黑色素瘤，也可视为高敏感和高特异的黑色素细胞标记。

Picture:	 Sample: Muscle (Mouse) Lysate at 40 ug Primary: Anti-MITF (SL1990R) at 1/1000 dilution Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution Predicted band size: 59 kD Observed band size: 57 kD