MITF 相关转录因子抗体

产品货号： mlR21815

英文名称： MITF

中文名称： MITF 相关转录因子抗体

别　　名： 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; MITF_HUMAN; WS2; WS2A.

研究领域： 细胞生物　免疫学　染色质和核信号　转录调节因子　表观遗传学

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应　： Human, Mouse, Rat, Chicken, Pig, Cow, Rabbit, Sheep,

产品应用： WB=1:500-2000 ELISA=1:500-1000

not yet tested in other applications.

optimal dilutions/concentrations should be determined by the end user.

分 子 量： 59kDa

细胞定位： 细胞核

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human MITF:1-100/526

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： 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

产品介绍： 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

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。

产品图片

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