

# Microphthalmia Transcription Factor (MITF) Antibody -With BSA and Azide

Mouse Monoclonal Antibody [Clone SPM290 ] Catalog # AH11834

# **Product Information**

Application	IHC, IF, FC
Primary Accession	<u>075030</u>
Other Accession	<u>4286, 166017, 618266</u>
Reactivity	Human
Host	Mouse
Clonality	Monoclonal
Isotype	Mouse / IgG1, kappa
Clone Names	SPM290
Calculated MW	58795

### **Additional Information**

Gene ID	4286
Other Names	Microphthalmia-associated transcription factor, Class E basic helix-loop-helix protein 32, bHLHe32, MITF, BHLHE32
Application Note	IHC~~1:100~500 IF~~1:50~200 FC~~1:10~50
Storage	Store at 2 to 8°C.Antibody is stable for 24 months.
Precautions	Microphthalmia Transcription Factor (MITF) Antibody - With BSA and Azide is for research use only and not for use in diagnostic or therapeutic procedures.

#### **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: <u>10587587</u> , PubMed: <u>22647378</u> , PubMed: <u>27889061</u> , PubMed: <u>9647758</u> ). 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: <u>10587587</u> , PubMed: <u>22647378</u> , PubMed: <u>27889061</u> , PubMed: <u>9647758</u> ).
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.

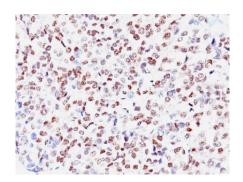
# Background

MITF (microphthalmia transcription factor) is a basic helix-loop-helix-leucine-zipper (bHLH-Zip) transcription factor that regulates the development and survival of melanocytes and retinal pigment epithelium, and also is involved in transcription of pigmentation enzyme genes such as tyrosinase TRP1 and TRP2. MITF has been shown to be phosphorylated by MAP kinase in response to c-kit activation, resulting in upregulation of MITF transcriptional activity. Mutations of the MITF gene are associated with the autosomal dominant hereditary deafness and pigmentation condition, Waardenburg Syndrome type 2A. Multiple isoforms of MITF exist, including MITF-A, MITF-B, MITF-C, MITF-H, and MITF-M, which differ in the amino-terminal domain and in their expression patterns. The MITF-M isoform is restricted to the melanocyte cell lineage. Anti-MITF, D5, recognizes a nuclear protein, which is expressed in the majority of primary and metastatic epithelioid malignant melanomas as well as in normal melanocytes, benign nevi and dysplastic nevi.

### References

Hemesath P, et. al. MAP kinase links the transcription factor microphthalmia to c-Kit signalling in melanocytes. Nature. 1998, 391:298-301 | Weilbaecher KN, et. al. Age-resolving osteopetrosis: a rat model implicating microphthalmia and the related transcription factor TFE3. J. Exp.Med. 1998, 187: 775-785 |

#### Images



Formalin-fixed, paraffin-embedded human Melanoma stained with MITF Monoclonal Antibody (SPM290).

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.