Best Tools for immunology Research

Total MITF Cell-Based Colorimetric ELISA Kit

Catalog No :	KA4180C
Reactivity :	Human;Mouse
Applications :	ELISA
Gene Name :	MITF
Human Gene Id :	4286
Human Swiss Prot No :	O75030
Mouse Swiss Prot No :	Q08874
Storage Stability :	2-8°C/6 months
Detection Method :	Colorimetric

Background : 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.,
Function :	eye development, regulation of myeloid leukocyte differentiation, transcription, regulation of transcription, DNA-dependent, regulation of transcription from RNA polymerase II promoter, cell surface receptor linked signal transduction, sensory organ development, positive regulation of biosynthetic process, positive regulation of macromolecule biosynthetic process, positive regulation of macromolecule metabolic process, positive regulation of gene expression, regulation of cell death, Wnt receptor signaling pathway, melanocyte differentiation, positive regulation of cellular biosynthetic process, regulation of cell proliferation, regulation of apoptosis, camera-type eye development, negative regulation of apoptosis, regulation of programmed cell death, negative regulation of programmed cell death, pigmentation, cell fate commitment, regulation of transcription, regulation of myeloid cell
Subcellular Location :	Nucleus . Cytoplasm . Found exclusively in the nucleus upon phosphorylation. .
Expression :	Expressed in melanocytes (at protein level). ; [Isoform A2]: Expressed in the retinal pigment epithelium, brain, and placenta (PubMed:9647758). Expressed in the kidney (PubMed:9647758, PubMed:10578055). ; [Isoform C2]: Expressed in the kidney and retinal pigment epithelium. ; [Isoform H1]: Expressed in the kidney. ; [Isoform H2]: Expressed in the kidney. ; [Isoform M1]: Expressed in melanocytes. ; [Isoform Mdel]: Expressed in melanocytes.
Sort :	22925
No4 :	1
Modifications :	Unmodified

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