Recombinant Human MITF, GST-tagged

Cat. No. MITF-30239TH **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant fragment with GST tag corresponding to amino acids 170-279 of Human MiTF; 110 amino acids, 38kDa.
Species	Human
Source	E.coli
ProteinLength	170-279 a.a.
Description	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.
Conjugation	GST
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 n
Form	Liquid

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Storage buffer	Preservative: NoneConstituents: 20% Glycerol, 50mM Tris acetate, 1mM EDTA, pH 7.5
Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.
Sequence Similarities	Belongs to the MiT/TFE family.Contains 1 basic helix-loop-helix (bHLH) domain.

GENE INFORMATION

Gene Name	MITF microphthalmia-associated transcription factor [Homo sapiens]
Official Symbol	MITF
Synonyms	MITF; microphthalmia-associated transcription factor; Waardenburg syndrome, type 2A , WS2, WS2A; bHLHe32; homolog of mouse microphthalmia; MI;
Gene ID	4286
mRNA Refseq	NM_000248
Protein Refseq	NP_000239
MIM	156845
Uniprot ID	O75030
Chromosome Location	3p14.1-p12.3
Pathway	IL6-mediated signaling events, organism-specific biosystem; Kit Receptor Signaling

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	Pathway, organism-specific biosystem; Melanogenesis, organism-specific biosystem; Melanogenesis, conserved biosystem; Melanoma, organism-specific biosystem;
Function	DNA binding; RNA polymerase II core promoter proximal region sequence-specific DNA binding transcription factor activity involved in positive regulation of transcription; protein binding;

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