abcam

Product datasheet

Recombinant Human SOX10 protein ab114238

1 Image

Description

Product name	Recombinant Human SOX10 protein
Expression system	Wheat germ
Accession	P56693
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	KPPGVALPTVSPPGVDAKAQVKTETAGPQGPPHYTDQP STSQIAYTSLSL PHYGSAFPSISRPQFDYSDHQPSGPYYGHSGQASGLYSA FSYMGPSQR
Predicted molecular weight	36 kDa including tags
Amino acids	336 to 433

Specifications

Our Abpromise guarantee covers the use of **ab114238** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	ELISA SDS-PAGE Western blot
Form	Liquid
Additional notes	

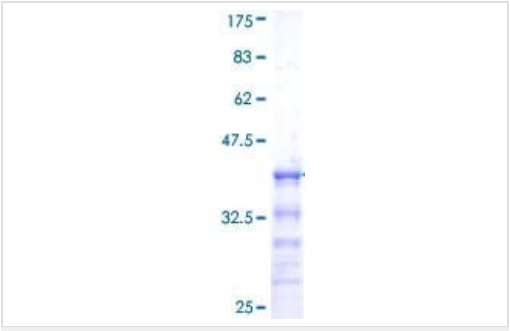
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.3% Glutathione, 0.79% Tris HCl

General Info

Function	Transcription factor that seems to function synergistically with the POU domain protein TST-1/OCT6/SCIP. Could confer cell specificity to the function of other transcription factors in developing and mature glia.
Tissue specificity	Expressed in fetal brain and in adult brain, heart, small intestine and colon.
Involvement in disease	Defects in SOX10 are the cause of Waardenburg syndrome type 2E (WS2E) [MIM:611584]. WS2 is a genetically heterogeneous, autosomal dominant disorder characterized by sensorineural deafness, pigmentary disturbances, and absence of dystopia canthorum. The frequency of deafness is higher in WS2 than in WS1. Defects in SOX10 are a cause of Waardenburg syndrome type 4C (WS4C) [MIM:613266]; also known as Waardenburg-Shah syndrome. WS4C is characterized by the association of Waardenburg features (depigmentation and deafness) and the absence of enteric ganglia in the distal part of the intestine (Hirschsprung disease). Defects in SOX10 are a cause of Yemenite deaf-blind hypopigmentation syndrome (YDBHS) [MIM:601706]. YDBHS consists of cutaneous hypopigmented and hyperpigmented spots and patches, microcornea, coloboma and severe hearing loss. Another case observed in a girl with similar skin symptoms and hearing loss but without microcornea or coloboma is reported as a mild form of this syndrome. Defects in SOX10 are the cause of peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease (PCWH) [MIM:609136]; also called neurologic variant of Waardenburg-Shah syndrome. PCWH is a rare, complex and more severe neurocristopathy that includes features of 4 distinct syndromes: peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease.
Sequence similarities	Contains 1 HMG box DNA-binding domain.
Cellular localization	Cytoplasm. Nucleus.

Images



SDS-PAGE - Recombinant Human SOX10 protein (ab114238)

12.5% SDS-PAGE showing ab114238 at approximately 36.41kDa stained with Coomassie Blue.

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