abcam

Product datasheet

Recombinant Human SOX10 protein ab114238

画像数 1

製品の詳細

製品名	Recombinant Human SOX10 protein
発現系	Wheat germ
アクセッション番号	P56693
タンパク質長	Protein fragment
Animal free	No
由来	Recombinant
生物種	Human
配列	KPPGVALPTVSPPGVDAKAQVKTETAGPQGPPHYTDQPSTSQIAYTSLSL PHYGSAFPSISRPQFDYSDHQPSGPYYGHSGQASGLYSAFSYMGPSQR
予測される分子量	36 kDa including tags
領域	336 to 433

特性

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.

アプリケーション	ELISA SDS-PAGE Western blot
製品の状態	Liquid

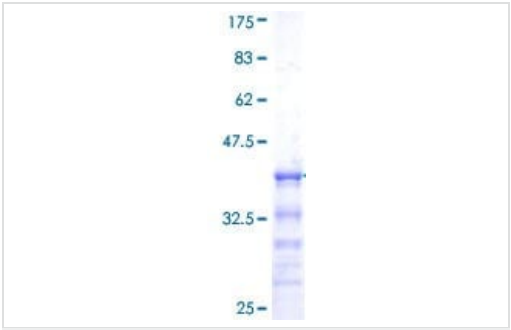
前処理および保存

保存方法および安定性	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

関連情報

機能	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.
組織特異性	Expressed in fetal brain and in adult brain, heart, small intestine and colon.
関連疾患	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.
配列類似性	Contains 1 HMG box DNA-binding domain.
細胞内局在	Cytoplasm. Nucleus.

画像



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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