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.

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| **Applications** | ELISA<br>SDS-PAGE<br>Western blot |
| **Form** | Liquid |
| **Additional notes** | |

### Preparation and Storage

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

## General Info

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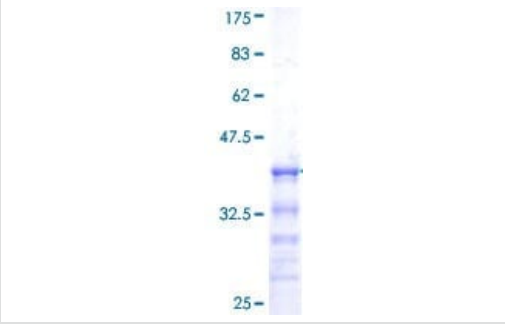
|---|---|
| **Function** | Transcription factor that seems to function synergistically with the POU domain protein TST-1/OCT6/SCIP. Could confer cell specificity to the function of other transcription factors in developing and mature glia. |
| **Tissue specificity** | Expressed in fetal brain and in 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.<br>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).<br>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.<br>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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