# abcam

## Product datasheet

## Recombinant Human SOX9 protein ab131911

1 References 1 Image

**Description** 

Product name Recombinant Human SOX9 protein

Expression system Wheat germ
Accession P48436

Protein length Full length protein

Animal free No

Nature Recombinant

**Species** Human

**Sequence** MNLLDPFMKMTDEQEKGLSGAPSPTMSEDSAGSPCPSG

**SGSDTENTRPQE** 

NTFPKGEPDLKKESEEDKFPVCIREAVSQVLKGYDWTLV

PMPVRVNGSSK

NKPHVKRPMNAFMVWAQAARRKLADQYPHLHNAELSKT

LGKLWRLLNESE

KRPFVEEAERLRVQHKKDHPDYKYQPRRRKSVKNGQAE

**AEEATEQTHISP** 

NAIFKALQADSPHSSSGMSEVHSPGEHSGQSQGPPTPPT

**TPKTDVQPGKA** 

DLKREGRPLPEGGRQPPIDFRDVDIGELSSDVISNIETFDV

NEFDQYLPP

NGHPGVPATHGQVTYTGSYGISSTAATPASAGHVWMSKQ

QAPPPPPQQPP

QAPPAPQAPPQPQAAPPQQPAAPPQQPQAHTLTTLSSE

**PGQSQRTHIKTE** 

QLSPSHYSEQQQHSPQQIAYSPFNLPHYSPSYPPITRSQY

**DYTDHQNSSS** 

YYSHAAGQGTGLYSTFTYMNPAQRPMYTPIADTSGVPSIPQ

THSPQHWEQ PVYTQLTRP

Predicted molecular weight 83 kDa including tags

Amino acids 1 to 509

Tags GST tag N-Terminus

**Specifications** 

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Our Abpromise guarantee covers the use of ab131911 in the following tested applications.

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

**Applications** ELISA

Western blot SDS-PAGE

Form Liquid

**Additional notes** 

#### **Preparation and Storage**

Stability and Storage Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.

pH: 8.00

Constituents: 0.3% Glutathione, 0.79% Tris HCI

#### **General Info**

**Function** Plays an important role in the normal skeletal development. May regulate the expression of other

genes involved in chondrogenesis by acting as a transcription factor for these genes.

**Involvement in disease** Defects in SOX9 are the cause of campomelic dysplasia (CMD1) [MIM:114290]. CMD1 is a rare,

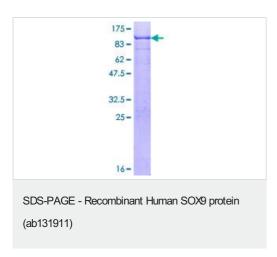
often lethal, dominantly inherited, congenital osteochondrodysplasia, associated with male-to-female autosomal sex reversal in two-thirds of the affected karyotypic males. A disease of the newborn characterized by congenital bowing and angulation of long bones, unusually small scapulae, deformed pelvis and spine and a missing pair of ribs. Craniofacial defects such as cleft palate, micrognatia, flat face and hypertelorism are common. Various defects of the ear are often evident, affecting the cochlea, malleus incus, stapes and tympanum. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial

cartilage and small thoracic cage.

Sequence similarities Contains 1 HMG box DNA-binding domain.

Cellular localization Nucleus.

## **Images**



ab131911 on a 12.5% SDS-PAGE Stained with Coomassie Blue.

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