# abcam

# Product datasheet

# Recombinant Human HSD3B2 protein ab114766

# 1 Image

**Description** 

Product name Recombinant Human HSD3B2 protein

Expression system Wheat germ
Accession P14060

Protein length Full length protein

Animal free No

Nature Recombinant

**Species** Human

Sequence MGWSCLVTGAGGLLGQRIVRLLVEEKELKEIRALDKAFRP

**ELREEFSKLQ** 

NRTKLTVLEGDILDEPFLKRACQDVSVVIHTACIIDVFGVTH

**RESIMNVN** 

VKGTQLLLEACVQASVPVFIYTSSIEVAGPNSYKEIIQNGHE

EEPLENTW

PTPYPYSKKLAEKAVLAANGWNLKNGDTLYTCALRPTYIYG

**EGGPFLSAS** 

INEALNNNGILSSVGKFSTVNPVYVGNVAWAHILALRALRD

**PKKAPSVRG** 

QFYYISDDTPHQSYDNLNYILSKEFGLRLDSRWSLPLTLMY

**WIGFLLEVV** 

SFLLSPIYSYQPPFNRHTVTLSNSVFTFSYKKAQRDLAYKP

LYSWEEAKQ KTVEWVGSLVDRHKETLKSKTQ

Predicted molecular weight 67 kDa including tags

Amino acids 1 to 372

#### **Specifications**

Our Abpromise guarantee covers the use of ab114766 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

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#### **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** 3-beta-HSD is a bifunctional enzyme, that catalyzes the oxidative conversion of Delta(5)-ene-3-

beta-hydroxy steroid, and the oxidative conversion of ketosteroids. The 3-beta-HSD enzymatic

system plays a crucial role in the biosynthesis of all classes of hormonal steroids.

Tissue specificity

Expressed in adrenal gland, testis and ovary.

**Pathway** 

Lipid metabolism; steroid biosynthesis.

Involvement in disease

Defects in HSD3B2 are the cause of adrenal hyperplasia type 2 (AH2) [MIM:201810]. AH2 is a form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late onset (NC or LOAH), and 'cryptic'

(asymptomatic). In AH2, virilization is much less marked or does not occur. AH2 is frequently

lethal in early life.

Note=Mild HSD3B2 deficiency in hyperandrogenic females is associated with characteristic traits of polycystic ovary syndrome, such as insulin resistance and luteinizing hormon hypersecretion.

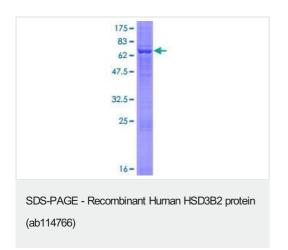
Sequence similarities

Belongs to the 3-beta-HSD family.

**Cellular localization** 

Endoplasmic reticulum membrane. Mitochondrion membrane.

### **Images**



12.5% SDS-PAGE Stained with Coomassie Blue showing ab114766 at approximately 66.99kDa.

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