abcam

Product datasheet

Recombinant Human NSDHL protein ab162360

1 Image

Description

Product name	Recombinant Human NSDHL protein
Expression system	Wheat germ
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MEPAVSEPMRDQVARTHLTEDTPKVNADIEKVNQNQAKR CTVIGGSGFLG QHMVEQLLARGYAVNVFDIQQGFDNPQVRFFLGDLCSRQ DLYPALKGVNT VFHCASPPPSSNNKELFYRVNYIGTKNVIETCKEAGVQKLI LTSSASVIF EGVDIKNGTEDLPYAMKPIDYYTETKILQERAVLGANDPEK NFLTTAIRP HGIFGPRDPQLVPILIEAARNGKMKFVIGNGKNLVDFTFVE NVVHGHILA AEQLSRDSTLGGKAFHITNDEPIPFWTFLSRILTGLNYEAPK YHIPYWVA YYLALLLSLLVMVISPVIQLQPTFTPMRVALAGTFHYYSCER AKKAMGYQ PLVTMDDAMERTVQSFRHLRRVK
Amino acids	1 to 373
Tags	GST tag N-Terminus

Specifications

Our **Abpromise guarantee** covers the use of **ab162360** 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
Form	Liquid
Additional notes	

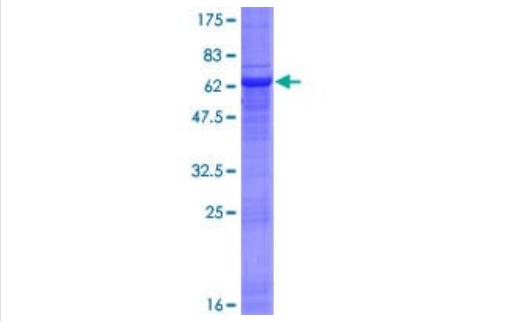
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Tissue specificity	Brain, heart, liver, lung, kidney, skin and placenta.
Pathway	Steroid biosynthesis; zymosterol biosynthesis; zymosterol from lanosterol: step 4/6.
Involvement in disease	Defects in NSDHL are the cause of congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD) [MIM:308050]. CHILD is an X-linked dominant disorder of lipid metabolism with disturbed cholesterol biosynthesis, which typically results in male lethality. Clinically, it is characterized by congenital, unilateral, ichthyosisform erythroderma with striking lateralization, sharp midline demarcation, and ipsilateral limb defects and hypoplasia of the body. Limbs defects range from hypoplasia of digits or ribs to complete amelia, often including scoliosis. Defects in NSDHL are the cause of CK syndrome (CKS) [MIM:300831]. CKS is a disorder characterized by mild to severe cognitive impairment, seizures, microcephaly, cerebral cortical malformations, dysmorphic facial features, and thin body habitus.
Sequence similarities	Belongs to the 3-beta-HSD family.
Cellular localization	Membrane.

Images



SDS-PAGE - Recombinant Human NSDHL protein (ab162360)

ab162360 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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- We provide support in Chinese, English, French, German, Japanese and Spanish
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