abcam

Product datasheet

Anti-NSDHL antibody ab54755

1 Image

Overview

Product name	Anti-NSDHL antibody
Description	Mouse monoclonal to NSDHL
Host species	Mouse
Tested applications	**Suitable for:** WB
Species reactivity	**Reacts with:** Recombinant fragment **Predicted to work with:** Human
Immunogen	Recombinant fragment, corresponding to amino acids 1-111 of Human NSDHL

Properties

Form	Liquid
Storage instructions	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.
Storage buffer	Preservative: None PBS, pH 7.2
Purity	Protein G purified
Clonality	Monoclonal
Isotype	IgG2a
Light chain type	kappa

Applications

Our Abpromise guarantee covers the use of **ab54755** in the following tested applications.

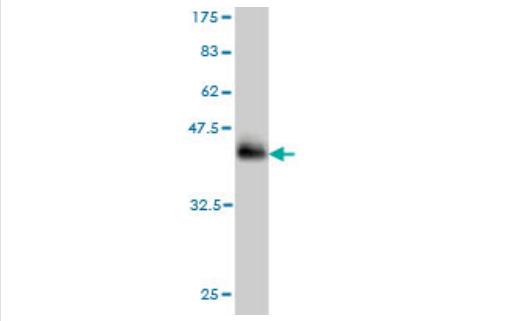
The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Application	Abreviews	Notes
WB		Use a concentration of 1 - 5 µg/ml. This antibody has only been tested in WB against the recombinant fragment used as immunogen. We have no data on the detection of endogenous protein.

Target

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



Western blot - Anti-NSDHL antibody (ab54755)

Western blot against tagged recombinant protein immunogen using ab54755 NSDHL antibody at 1ug/ml. Predicted band size of immunogen is 38 kDa.

This antibody has only been tested in WB against the recombinant fragment used as immunogen. We have no data on the detection of endogenous protein.

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