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NSDHL Rabbit mAb

Catalog # AP76625

Product Information

Application	WB, IP
Primary Accession	Q15738
Reactivity	Rat, Human, Mouse
Host	Rabbit
Clonality	Monoclonal Antibody
Isotype	IgG
Conjugate	Unconjugated
Purification	Affinity Purified
Calculated MW	41900

Additional Information

Gene ID	50814
Other Names	NSDHL
Dilution	WB~~1:1000 IP~~N/A
Format	Liquid in 50mM Tris-Glycine(pH 7.4), 0.15M NaCl, 40%Glycerol, 0.01% sodium azide and 0.05% BSA.
Storage	Store at 4°C short term. Aliquot and store at -20°C long term. Avoid freeze/thaw cycles.

Protein Information

Name	NSDHL
Synonyms	H105E3
Function	Catalyzes the NAD(P)(+)-dependent oxidative decarboxylation of the C4 methyl groups of 4-alpha-carboxysterols in post-squalene cholesterol biosynthesis (By similarity). Also plays a role in the regulation of the endocytic trafficking of EGFR (By similarity).
Cellular Location	Endoplasmic reticulum membrane; Single-pass membrane protein. Lipid droplet {ECO:0000250|UniProtKB:Q9R1J0} Note=Trafficking through the Golgi is necessary for ER membrane localization. {ECO:0000250|UniProtKB:Q9R1J0}
Tissue Location	Brain, heart, liver, lung, kidney, skin and placenta

Background

The protein encoded by this gene is localized in the endoplasmic reticulum and is involved in cholesterol biosynthesis. Mutations in this gene are associated with CHILD syndrome, which is a X-linked dominant disorder of lipid metabolism with disturbed cholesterol biosynthesis, and typically lethal in males. Alternatively spliced transcript variants with differing 5' UTR have been found for this gene.

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