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DHCR7 Rabbit pAb

Catalog No.: A8049 **1 Publications**

Basic Information

Observed MW
70kDa

Calculated MW
54kDa

Category
Primary antibody

Applications
ELISA,WB

Cross-Reactivity
Human

Background

This gene encodes an enzyme that removes the C(7-8) double bond in the B ring of sterols and catalyzes the conversion of 7-dehydrocholesterol to cholesterol. This gene is ubiquitously expressed and its transmembrane protein localizes to the endoplasmic reticulum membrane and nuclear outer membrane. Mutations in this gene cause Smith-Lemli-Opitz syndrome (SLOS); a syndrome that is metabolically characterized by reduced serum cholesterol levels and elevated serum 7-dehydrocholesterol levels and phenotypically characterized by cognitive disability, facial dysmorphism, syndactyly of second and third toes, and holoprosencephaly in severe cases to minimal physical abnormalities and near-normal intelligence in mild cases. Alternative splicing results in multiple transcript variants that encode the same protein.

Recommended Dilutions

WB	1:500 - 1:2000

Immunogen Information

Gene ID
1717

Swiss Prot
Q9UBM7

Immunogen
Recombinant fusion protein containing a sequence corresponding to amino acids 346-475 of human DHCR7 (NP_001351.2).

Synonyms
SLOS; DHCR7

Contact

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Product Information

Source
Rabbit

Isotype
IgG

Purification
Affinity purification

Storage
Store at -20℃. Avoid freeze / thaw cycles.
Buffer: PBS with 0.05% proclin300,50% glycerol,pH7.3.