DHCR7 Rabbit Polyclonal Antibody

CAB8049

Product Information

Size:

20uL, 50uL, 100uL, 200uL

Observed MW:

70kDa

Calculated MW:

54kDa

Applications:

WB

Reactivity:

Human

Antibody Information

Recommended dilutions:
WB 1:500 - 1:2000

Source:
Rabbit

Isotype:
IgG

Purification:
Affinity purification

Protein 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 mental retardation, 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.

Immunogen information

Gene ID:
1717

Uniprot
Q9UBM7

Synonyms:
DHCR7; SLOS

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

Storage:
Store at -20℃. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

Product Images

