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DHCR7

Reactivity: Human

Tested applications:WB IHC IF

Recommended Dilution:WB 1:500 - 1:2000 IHC 1:50 - 1:200 IF 1:50 - 1:200

Calculated MW:54kDa

Observed MW: Refer to figures

Immunogen:

Recombinant protein of human DHCR7

Storage Buffer:

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

Synonym:

SLOS;

Catalog #:A8049

Antibody Type:

Polyclonal Antibody

Species: Rabbit

Gene ID:1717

Isotype: IgG

Swiss Prot:Q9UBM7

Purity: Affinity purification

For research use only.

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.

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