

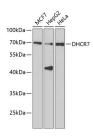
Tel:240-252-7368(USA) Fax: 240-252-7376(USA) techsupport@elabscience.com Website: www.elabscience.com

DHCR7 Polyclonal Antibody

Catalog No. E-AB-67941 Reactivity Η Store at -20°C. Avoid freeze / thaw cycles. Rabbit **Storage** Host **Applications Isotype IgG**

Note: Centrifuge before opening to ensure complete recovery of vial contents.

Images



Western blot analysis of extracts of various cell lines using DHCR7 Polyclonal Antibody at dilution of 1:1000.

Immunogen Information

Immunogen Recombinant fusion protein of human DHCR7

(NP_001351.2).

GeneID 1717 **Swissprot** Q9UBM7 DHCR7,SLOS **Synonyms**

Product Information

Calculated MW 54kDa **Observed MW** 70kDa

Buffer PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

Purify Affinity purification Dilution WB 1:500-1:2000

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.