

DHCR7 Antibody (C-term) Blocking peptide

Synthetic peptide Catalog # BP11452b

Specification

DHCR7 Antibody (C-term) Blocking peptide - Product Information

Primary Accession

<u>Q9UBM7</u>

DHCR7 Antibody (C-term) Blocking peptide - Additional Information

Gene ID 1717

Other Names 7-dehydrocholesterol reductase, 7-DHC reductase, Putative sterol reductase SR-2, Sterol Delta(7)-reductase, DHCR7, D7SR

Format

Peptides are lyophilized in a solid powder format. Peptides can be reconstituted in solution using the appropriate buffer as needed.

Storage Maintain refrigerated at 2-8°C for up to 6 months. For long term storage store at -20°C.

Precautions This product is for research use only. Not for use in diagnostic or therapeutic procedures.

DHCR7 Antibody (C-term) Blocking peptide - Protein Information

Name DHCR7

Synonyms D7SR

Function

7-dehydrocholesterol reductase of the cholesterol biosynthetic pathway reducing the C7-C8 double bond of cholesta-5,7- dien-3beta-ol (7-dehydrocholesterol/7-DHC) and cholesta-5,7,24-trien-3beta-ol, two intermediates in that pathway.

Cellular Location Endoplasmic reticulum membrane; Multi-pass membrane protein

Tissue Location Widely expressed. Most abundant in adrenal gland, liver, testis, and brain.

DHCR7 Antibody (C-term) Blocking peptide - Protocols

Provided below are standard protocols that you may find useful for product applications.



<u>Blocking Peptides</u>

DHCR7 Antibody (C-term) Blocking peptide - Images

DHCR7 Antibody (C-term) Blocking peptide - Background

This gene encodes an enzyme that removes the C(7-8) doublebond in the B ring of sterols and catalyzes the conversion of7-dehydrocholesterol to cholesterol. This gene is ubiquitouslyexpressed and its transmembrane protein localizes to theendoplasmic reticulum membrane and nuclear outer membrane. Mutations in this gene cause Smith-Lemli-Opitz syndrome (SLOS); asyndrome that is metabolically characterized by reduced serumcholesterol levels and elevated serum 7-dehydrocholesterol levelsand phenotypically characterized by mental retardation, facialdysmorphism, syndactyly of second and third toes, andholoprosencephaly in severe cases to minimal physical abnormalities near-normal intelligence in mild cases. Alternative splicingresults in multiple transcript variants that encode the sameprotein.

DHCR7 Antibody (C-term) Blocking peptide - References

Bailey, S.D., et al. Diabetes Care 33(10):2250-2253(2010)Koo, G., et al. Am. J. Med. Genet. A 152A (8), 2094-2098 (2010) :Wang, T.J., et al. Lancet 376(9736):180-188(2010)Ahn, J., et al. Hum. Mol. Genet. 19(13):2739-2745(2010)Jugessur, A., et al. PLoS ONE 5 (7), E11493 (2010) :