

Recombinant Human DHCR7 Protein, GST-tagged

Cat. No. DHCR7-2582H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human DHCR7 full-length ORF (NP_001351.1, 1 a.a. - 475 a.a.) recombinant protein with GST-tag at N-terminal.
Species	Human
Source	Wheat Germ
Description	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.[provided by RefSeq, Aug 2009]
Molecular Mass	80.9 kDa
AA Sequence	MAAKLQPNIPKAKSLDGVTDNDRTASQGQWGRAWVDWFSLASVIFLLLFAPFIVYYF IMACDQYSCALTGPVVDIVTGHARLSDIWAKTPPITRKAAQLYTLWVTFQVLLYTSLP DFCHKFLPGYVGGIQEGAVTPAGVVNKKYQINGLQAWLLTHLLWFANAHLLSWFSPTI

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IFDNWIPLLWCANILGYAVSTFAMVKG YFFPTSARDCKFTGNFFYNYMMGIEFNPRIG
 KWFDKLFNFNGRPGIVAWTLINLSFAAKQRELHSHVTNAMVLNVNLQAIYVIDFFWNE
 TWYLKTIDICHDFGWYLGWDCVWLPYLYTLQGLYLVYHPVQLSTPHAVGVLLLG
 LVGYIIFRVANHQKDLFRRTDGRCLIWGRKPKVIECSYTSADGQRHHSKLLVSGFW
 GVARHFNYVGDLMGSLAYCLACGGGHLLPYFYIIMAILLTHRCLRDEHRCASKYGR
 DWERYTAAVPYRLLPGIF

Applications

Enzyme-linked Immunoabsorbent Assay
 Western Blot (Recombinant protein)
 Antibody Production
 Protein Array

Notes

Best use within three months from the date of receipt of this protein.

Storage

Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.

Storage Buffer

50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

GENE INFORMATION

Gene Name

DHCR7 7-dehydrocholesterol reductase [Homo sapiens]

Official Symbol

DHCR7

Synonyms

DHCR7; 7-dehydrocholesterol reductase; SLOS, Smith Lemli Opitz syndrome; 7-DHC reductase; sterol delta-7-reductase; putative sterol reductase SR-2; delta-7-dehydrocholesterol reductase; SLOS;

Gene ID

1717

mRNA Refseq

NM_001163817

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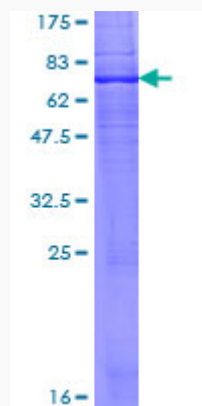
 45-1 Ramsey Road, Shirley, NY 11967, USA

Protein Refseq NP_001157289

MIM 602858

UniProt ID Q9UBM7

**Quality Control
Testing:**



12.5% SDS-PAGE Stained with Coomassie Blue.

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