

Recombinant Human DHCR7 Protein, His/GST-tagged

Cat. No. DHCR7-479H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human DHCR7(Cys61~Leu153) fused with His/GST tag at N-terminal was expressed in E. coli.
Species	Human
Source	E.coli
ProteinLength	Cys61~Leu153
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.
Form	20mM Tris, 150mM NaCl, pH8.0, containing 1mM EDTA, 1mM DTT, 0.01% SKL, 5% Trehalose and Proclin300.
Molecular Mass	40.2kDa

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Endotoxin	<1.0EU per 1ug (determined by the LAL method)
Purity	> 95%
Applications	Positive Control; Immunogen; SDS-PAGE; WB. If bio-activity of the protein is needed, please check active protein
Stability	The thermal stability is described by the loss rate. The loss rate was determined by accelerated thermal degradation test, that is, incubate the protein at 37 centigrade for 48h, and no obvious degradation and precipitation were observed. The loss rate is less than 5% within the expiration date under appropriate storage condition.
Storage	Avoid repeated freeze/thaw cycles. Store at 2-8 centigrade for one month. Aliquot and store at -80 centigrade for 12 months.
Reconstitution	Reconstitute in 20mM Tris, 150mM NaCl (pH8.0) to a concentration of 0.1-1.0 mg/mL. Do not vortex.

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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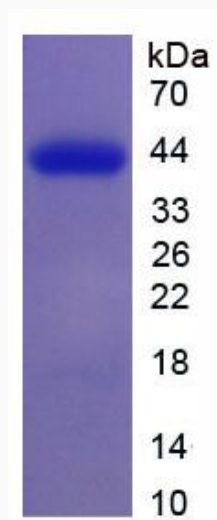
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Protein Refseq NP_001157289

MIM 602858

UniProt ID Q9UBM7



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