

Product Information

MemDX™ Membrane Protein Human DHCR7 (7-dehydrocholesterol reductase) Expressed *in vitro* E.coli expression system, Full Length

Cat. No.: **MPX3602K**

This product is for research use only and is not intended for diagnostic use.

This product is a Human DHCR7 membrane protein expressed *in vitro* E.coli expression system. The protein is for research use only and is not approved for use in humans or in clinical diagnosis.

Product Specifications

Host Species

Human

Target Protein

DHCR7

Protein Length

Full Length

Protein Class

Oxidoreductase

TMD

7

Sequence

MAAKSQPNIPKAKSLDGVTNDRTASQGQWGRAWVDWFLASVIFLLLFAPFIVYYFIMACDQYSCALTGPVVDIVTG HARLSDIWA

Product Description

Expression Systems

in vitro E.coli expression system

Tag

10xHis tag at the N-terminus

Protein Format

Soluble

Form

Liquid or Lyophilized powder

Buffer

Tris/PBS-based buffer, 6% Trehalose, pH 8.0

Storage

Aliquot and store at -20°C or lower. For long term storage, we recommend to store at -70°C or lower. Avoid freeze/thaw cycles.

Target

Target Protein

DHCR7

Full Name

7-dehydrocholesterol reductase

Introduction

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.

Alternative Names

DHCR7; SLOS; 7-DHC reductase; delta-7-dehydrocholesterol reductase; delta7-sterol reductase; putative sterol reductase SR-2; sterol delta-7-reductase; sterol reductase SR-2; 7-dehydrocholesterol reductase

Gene ID

[1717](#)

UniProt ID

[Q9UBM7](#)