

## 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

MAAKSQPNIPKAKSLDGVTNDRTASQQQWGRAWEVDWFLASVIFLLFAPFIVYYFIMACDQYSCALTGPVVDIVTGHARLSDIWA

#### 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](#)