

## DHCR7 抗原（重组蛋白）

中文名称： DHCR7 抗原（重组蛋白）

英文名称： DHCR7 Antigen (Recombinant Protein)

别名： SLOS

储存： 冷冻（-20℃）

相关类别： 抗原

概述：

Fusion protein corresponding to a region derived from 61-153 amino acids of human DHCR7

技术规格：

<b>Full name:</b>	7-dehydrocholesterol reductase
<b>Synonyms:</b>	SLOS
<b>Swissprot:</b>	Q9UBM7
<b>Gene Accession:</b>	BC000054
<b>Purity:</b>	>85%, as determined by Coomassie blue stained SDS-PAGE
<b>Expression system:</b>	Escherichia coli
<b>Tags:</b>	His tag C-Terminus, GST tag N-Terminus
<b>Background:</b>	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 mental retardation, 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.