Product Datasheet

Recombinant Human DHCR7

Catalog No: #GP11461

Package Size: #GP11461-1 100ug

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Description

Product Name	Recombinant Human DHCR7
Brief Description	Recombinant Protein
Immunogen Description	Fusion protein corresponding to a region derived from 61-153 amino acids of human 7-dehydrocholesterol reductase
Target Name	7-dehydrocholesterol reductase
Other Names	SLOS
Accession No.	Swissprot:Q9UBM7Gene Accession:BC000054
Uniprot	Q9UBM7
GeneID	1717;
Storage	-20~-80°C, pH 7.6 PBS

Background

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.

References

Note: For in vitro research use only, not for diagnostic or therapeutic use. This product is not a medical device.

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