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Product datasheet for RC200480L2V

DHCR7 (NM_001360) Human Tagged ORF Clone Lentiviral Particle

Product data:

Product Type:	Lentiviral Particles
Product Name:	DHCR7 (NM_001360) Human Tagged ORF Clone Lentiviral Particle
Symbol:	DHCR7
Synonyms:	SLOS
Mammalian Cell Selection:	None
Vector:	pLenti-C-mGFP (PS100071)
Tag:	mGFP
ACCN:	NM_001360
ORF Size:	1425 bp
ORF Nucleotide Sequence:	The ORF insert of this clone is exactly the same as(RC200480).
OTI Disclaimer:	The molecular sequence of this clone aligns with the gene accession number as a point of reference only. However, individual transcript sequences of the same gene can differ through naturally occurring variations (e.g. polymorphisms), each with its own valid existence. This clone is substantially in agreement with the reference, but a complete review of all prevailing variants is recommended prior to use. More info
OTI Annotation:	This clone was engineered to express the complete ORF with an expression tag. Expression varies depending on the nature of the gene.
RefSeq:	NM_001360.2
RefSeq Size:	2665 bp
RefSeq ORF:	1428 bp
Locus ID:	1717
UniProt ID:	Q9UBM7
Cytogenetics:	11q13.4
Domains:	ERG4_ERG24
Protein Families:	Druggable Genome, Transmembrane

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Protein Pathways: Metabolic pathways, Steroid biosynthesis

MW: 54.5 kDa

Gene Summary: 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.[provided by RefSeq, Aug 2009]