

## DHCR7 Antibody (C-term)

Catalog_no :	AB0924
Applications :	WB, IHC-P, FC
Reactivity :	H, M
Category :	抗原抗体
Size :	100 $\mu$ L/50 $\mu$ L
Immunogen :	HUMAN:437-463
Specificity :	This DHCR7 antibody is generated from rabbits immunized with a KLH conjugated synthetic peptide between 437-463 amino acids from the C-terminal region of human DHCR7.
Dilution :	IF,1:25;WB,1:2000;WB,1:1000;IHC-P,1:50~100;
Purification :	Purified polyclonal antibody supplied in PBS with 0.09% (W/V) sodium azide. This antibody is purified through a protein A column, followed by peptide affinity purification.
Other_name :	7-dehydrocholesterol reductase, 7-DHC reductase, Putative sterol reductase SR-2, Sterol Delta(7)-reductase, DHCR7, D7SR
Isotype :	Rabbit Ig
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.
reference :	Bailey, S.D., et al. Diabetes Care 33(10):2250-2253(2010) Koo, G., et al. Am. J. Med. Genet. A 152A (8), 2094-2098 (2010) : Wang, T.J., et al. Lancet 376(9736):180-188(2010) Ahn, J., et al. Hum. Mol. Genet. 19(13):2739-2745(2010) Jugessur, A., et al.