

Product datasheet for **TA361232**

DHCR7 Rabbit Polyclonal Antibody

Product data:

Product Type:	Primary Antibodies
Applications:	WB
Reactivity:	Human
Host:	Rabbit
Clonality:	Polyclonal
Immunogen:	The immunogen is a synthetic peptide directed towards the N terminal region of human DHCR7
Specificity:	Expected reactivity: Human
Formulation:	Liquid. Purified antibody supplied in 1x PBS buffer with 0.09% (w/v) sodium azide and 2% sucrose. <i>Note that this product is shipped as lyophilized powder to China customers.</i>
Concentration:	lot specific
Purification:	Affinity purified
Conjugation:	Unconjugated
Storage:	For short term use, store at 2-8°C up to 1 week. For long term storage, store at -20°C in small aliquots to prevent freeze-thaw cycles.
Stability:	Shelf life: one year from despatch.
Predicted Protein Size:	52 kDa
Gene Name:	7-dehydrocholesterol reductase
Database Link:	NP_001157289.1 Entrez Gene 1717 Human Q9UBM7



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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.

Synonyms:

D7SR; SLOS

Protein Families:

Druggable Genome, Transmembrane

Protein Pathways:

Metabolic pathways, Steroid biosynthesis

Product images: