10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

Rabbit Anti-DHCR7 antibody

Purified Rabbit Polyclonal Antibody (Pab)
Catalog # AP50932

Product Information

Application	WB, E
Primary Accession	Q9UBM7
Reactivity	Human, Mouse, Rat, Chicken, Dog, Horse, Bovine
Host	Rabbit
Clonality	polyclonal
Calculated MW	54489
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human DHCR7
Epitope Specificity	351-450/475
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Endoplasmic reticulum membrane; Multi-pass membrane protein.
SIMILARITY	Belongs to the ERG4/ERG24 family.
DISEASE	Defects in DHCR7 are the cause of Smith-Lemli-Opitz syndrome (SLOS) [MIM:270400]; also known as SLO syndrome or RSH syndrome. SLOS is an autosomal recessive frequent inborn disorder of sterol metabolism with characteristic congenital malformations and dysmorphias. All patients suffer from mental retardation. Children with SLOS have elevated serum 7-dehydrocholesterol (7-DHC) levels and low serum cholesterol levels. SLOS occurs in relatively high frequency: approximately 1 in 20,000 to 30,000 births in populations of northern and central European background. Historically, a clinical distinction often was made between classic ('type I') SLOS and the more severely affected ('type II') patients. There is, in reality, a clinical and biochemical continuum from mild to severe SLOS.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	The DHCR7 gene encodes delta-7-sterol reductase (EC 1.3.1.21), the penultimate enzyme of mammalian sterol biosynthesis that converts 7-dehydrocholesterol (7-DHC) to cholesterol.

Additional Information

Gene ID	1717
Other Names	7-dehydrocholesterol reductase, 7-DHC reductase, Putative sterol reductase SR-2, Sterol Delta(7)-reductase, DHCR7, D7SR
Target/Specificity	Most abundant in adrenal gland, liver, testis, and brain.
Dilution	WB=1:500-2000,ELISA=1:5000-10000

Format	0.01M TBS(pH7.4) with 1% BSA, 0.09% (W/V) sodium azide and 50% Glyce
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	DHCR7 (HGNC:2860)
Synonyms	D7SR
Function	Oxidoreductase that catalyzes the last step of the cholesterol synthesis pathway, which transforms cholesta-5,7-dien- 3beta-ol (7-dehydrocholesterol,7-DHC) into cholesterol by reducing the C7-C8 double bond of its sterol core (PubMed:25637936, PubMed:38297129, PubMed:38297130, PubMed:9465114, PubMed:9634533). Can also metabolize cholesta-5,7,24-trien-3beta-ol (7-dehydrodemosterol, 7-DHD) to desmosterol, which is then metabolized by the Delta(24)-sterol reductase (DHCR24) to cholesterol (By similarity). Modulates ferroptosis (a form of regulated cell death driven by iron-dependent lipid peroxidation) through the metabolic breakdown of the anti- ferroptotic metabolites 7-DHC and 7-DHD which, when accumulated, divert the propagation of peroxyl radical-mediated damage from phospholipid components to its sterol core, protecting plasma and mitochondrial membranes from phospholipid autoxidation (PubMed:38297129, PubMed:38297130).
Cellular Location	Endoplasmic reticulum membrane; Multi-pass membrane protein
Tissue Location	Widely expressed. Most abundant in adrenal gland, liver, testis, and brain.

Background

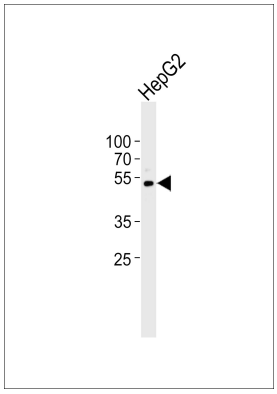
Production of cholesterol by reduction of C7-C8 double bond of 7-dehydrocholesterol (7-DHC).

References

Waterham H.R.,et al.Am. J. Hum. Genet. 63:329-338(1998).
Moebius F.F.,et al.Proc. Natl. Acad. Sci. U.S.A. 95:1899-1902(1998).
Holmer L.,et al.Genomics 54:469-476(1998).
Ota T.,et al.Nat. Genet. 36:40-45(2004).
Wassif C.A.,et al.Am. J. Hum. Genet. 63:55-62(1998).

Images

Western blot analysis of lysate from HepG2 cell line, using Rabbit Anti-DHCR7 antibody(AP50932). AP50932 was diluted at 1:1000. A goat anti-rabbit IgG H&L(HRP) at 1:10000 dilution was used as the secondary antibody.Lysate at 20ug.



Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.