AKR1D1 Peptide - Middle Region Blocking Peptide

PRODUCT OVERVIEW

Product Name	AKR1D1, Blocking Peptide
Full Product Name	AKR1D1 Peptide - middle region
Product Gene Name	AKR1D1 blocking peptide
Product Synonym Gene Name	CBAS2; SRD5B1; 3o5bred
Sequence	Synthetic peptide located within the following region: RVLQLDYVDL YIIEVPMAFK PGDEIYPRDE NGKWLYHKSN LCATWEAMEA
Species Reactivity	Human
Form/Format	Lyophilized powder
Preparation and Storage	Add 100ul of sterile PBS. Final peptide concentration is 1 mg/ml in PBS. For longer periods of storage, store at -20 degree C. Avoid repeat freeze-thaw cycles.
Antibody/Peptide Pairs	AKR1D1 peptide (MBS3246574) is used for blocking the activity of AKR1D1 antibody (MBS3221848)
Molecular Weight	35 kDa
OMIM	235555
Product Categories/Family for AKR1D1 blocking peptide	Peptide
Related Product Information for AKR1D1 blocking peptide	This is a synthetic peptide designed for use in combination with anti- AKR1D1 Antibody, made Target Description: The enzyme encoded by this gene is responsible for the catalysis of the 5-beta-reduction of bile acid intermediates and steroid hormones carrying a delta(4)-3-one structure. Deficiency of this enzyme may contribute to hepatic dysfunction. Three transcript variants encoding different isoforms have been found for this gene. Other variants may be present, but their full-length natures have not been determined yet.
Other Notes	Small volumes of AKR1D1 blocking peptide vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.
Searchable Terms for AKR1D1 purchase	MBS3246574 is a blocking peptide targeting AKR1D1. Also known as AKR1D1 Peptide - middle region. Reactive in Human.

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for AKR1D1. It may not necessarily be applicable to this product.

NCBI GI #	300116271
NCBI GeneID	6718
NCBI Accession #	NP_001177835.1
UniProt Primary Accession #	P51857
UniProt Related Accession #	P51857
GenBank Nucleotide #	NM_001190906.1
NCBI Official Full Name	3-oxo-5-beta-steroid 4-dehydrogenase isoform 2
NCBI Official Synonym Full Names	aldo-keto reductase family 1 member D1

NCBI Official Symbol	AKR1D1
NCBI Official Synonym Symbols	CBAS2; SRD5B1; 3o5bred
UniProt Protein Name	3-oxo-5-beta-steroid 4-dehydrogenase
UniProt Synonym Protein Names	Aldo-keto reductase family 1 member D1; Delta(4)-3-ketosteroid 5-beta-reductase; Delta(4)-3-oxosteroid 5-beta-reductase
Protein Family	3-oxo-5-beta-steroid 4-dehydrogenase
UniProt Gene Name	AKR1D1
UniProt Synonym Gene Names	SRD5B1
UniProt Entry Name	AK1D1_HUMAN
NCBI Protein Information	3-oxo-5-beta-steroid 4-dehydrogenase
NCBI Summary for AKR1D1	The enzyme encoded by this gene is responsible for the catalysis of the 5-beta-reduction of bile acid intermediates and steroid hormones carrying a delta(4)-3-one structure. Deficiency of this enzyme may contribute to hepatic dysfunction. Three transcript variants encoding different isoforms have been found for this gene. Other variants may be present, but their full-length natures have not been determined yet. [provided by RefSeq, Jul 2010]
UniProt Comments for AKR1D1	AKR1D1: Efficiently catalyzes the reduction of progesterone, androstenedione, 17-alpha-hydroxyprogesterone and testosterone to 5-beta-reduced metabolites. The bile acid intermediates 7- alpha,12-alpha-dihydroxy-4-cholesten-3-one and 7-alpha-hydroxy-4- cholesten-3-one can also act as substrates. Defects in AKR1D1 are the cause of congenital bile acid synthesis defect type 2 (CBAS2); also known as cholestasis with delta(4)-3-oxosteroid 5-beta-reductase deficiency. Patients with this liver disease show absence or low levels of chenodeoxycholic acid and cholic acid in plasma and urine. Belongs to the aldo/keto reductase family. 3 isoforms of the human protein are produced by alternative splicing. Protein type: EC 1.3.1.3; Lipid Metabolism - C21-steroid hormone; Lipid Metabolism - androgen and estrogen; Lipid Metabolism - primary bile acid biosynthesis; Oxidoreductase Chromosomal Location of Human Ortholog: 7q32-q33 Cellular Component: cytosol Molecular Function: aldo-keto reductase activity; steroid binding Biological Process: androgen metabolic process; bile acid biosynthetic process; C21-steroid hormone metabolic process; cholesterol catabolic process; digestion Disease: Bile Acid Synthesis Defect, Congenital, 2
Research Articles on AKR1D1	**1.** infant proved to be a compound heterozygote of the AKR1D1 variants c.579+2delT and c.853C>T(p.Q285X), two novel mutations originated from his mother and father, respectively
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