NPHP4 Peptide - N-Terminal Region Blocking Peptide

PRODUCT OVERVIEW

Product Name	NPHP4, Blocking Peptide
Full Product Name	NPHP4 Peptide - N-terminal region
Product Gene Name	NPHP4 blocking peptide
Product Synonym Gene Name	NPHP4;KIAA0673;
Sequence	Synthetic peptide located within the following region: VSGPKVERRP SRKPPTSPSS PPAPVPRVLA APQNSPVGPG LSISQLAASP
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	NPHP4 peptide (MBS3244973) is used for blocking the activity of NPHP4 antibody (MBS3220150)
Molecular Weight	156kDa
OMIM	606966
Product Categories/Family for NPHP4 blocking peptide	Peptide
Related Product Information for NPHP4 blocking peptide	This is a synthetic peptide designed for use in combination with anti-NPHP4 Antibody, made Target Description: This gene encodes a protein involved in renal tubular development and function. This protein interacts with nephrocystin, and belongs to a multifunctional complex that is localized to actin- and microtubule-based structures. Mutations in this gene are associated with nephronophthisis type 4, a renal disease, and with Senior-Loken syndrome type 4, a combination of nephronophthisis and retinitis pigmentosa. Alternative splicing results in multiple transcript variants.
Other Notes	Small volumes of NPHP4 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 NPHP4 purchase	MBS3244973 is a blocking peptide targeting NPHP4. Also known as NPHP4 Peptide - N-terminal region. Reactive in Human.

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for NPHP4. It may not necessarily be applicable to this product.

NCBI GI #	578798909
NCBI GeneID	261734
NCBI Accession #	XP_006710628
UniProt Primary Accession #	O75161
UniProt Related Accession #	O75161
GenBank Nucleotide #	XM_006710565.2
NCBI Official Synonym Full Names	nephrocystin 4

NCBI Official Symbol	NPHP4
NCBI Official Synonym Symbols	POC10; SLSN4
UniProt Protein Name	Nephrocystin-4
UniProt Synonym Protein Names	Nephroretinin
Protein Family	Nephrocystin
UniProt Gene Name	NPHP4
UniProt Synonym Gene Names	KIAA0673
UniProt Entry Name	NPHP4_HUMAN
NCBI Protein Information	nephrocystin-4
NCBI Summary for NPHP4	This gene encodes a protein involved in renal tubular development and function. This protein interacts with nephrocystin, and belongs to a multifunctional complex that is localized to actin- and microtubule-based structures. Mutations in this gene are associated with nephronophthisis type 4, a renal disease, and with Senior-Loken syndrome type 4, a combination of nephronophthisis and retinitis pigmentosa. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Apr 2014]
UniProt Comments for NPHP4	NPHP4: Involved in the organization of apical junctions in kidney cells together with NPHP1 and RPGRIP1L/NPHP8. Does not seem to be strictly required for ciliogenesis. Defects in NPHP4 are the cause of nephronophthisis type 4 (NPHP4); also known as familial juvenile nephronophthisis 4. NPHP4 is an autosomal recessive inherited disease resulting in end-stage renal disease at age ranging between 6 and 35 years. It is a progressive tubulo-interstitial kidney disorder characterized by polydipsia, polyuria, anemia and growth retardation. The most prominent histological features are modifications of the tubules with thickening of the basement membrane, interstitial fibrosis and, in the advanced stages, medullary cysts. Ciliary dysfunction leads to a broad spectrum of disorders, collectively termed ciliopathies. Overlapping clinical features include retinal degeneration, renal cystic disease, skeletal abnormalities, fibrosis of various organ, and a complex range of anatomical and functional defects of the central and peripheral nervous system. The ciliopathy range of diseases includes Meckel-Gruber syndrome, Bardet-Biedl syndrome, Joubert syndrome, nephronophtisis, Senior-Loken syndrome, and Jeune asphyxiating thoracic dystrophy among others. Single-locus allelism is insufficient to explain the variable penetrance and expressivity of such disorders, leading to the suggestion that variations across multiple sites of the ciliary proteome, including NPHP4, influence the clinical outcome. Defects in NPHP4 are the cause of Senior-Loken syndrome type 4 (SLSN4). SLSN is a renal-retinal disorder characterized by progressive wasting of the filtering unit of the kidney, with or without medullary cystic renal disease, and progressive eye disease. Typically this disorder becomes apparent during the first year of life. Belongs to the NPHP4 family. Protein type: Cytoskeletal Chromosomal Location of Human Ortholog: 1p36 Cellular Component: centrosome; tight junction; membrane; intercellular junction; cytosol; photoreceptor connecting cilium Molecular Function: protein binding; structural molecule activity Biological Process: sperm motility; cell-cell adhesion; retina development in camera-type eye; organelle organization and biogenesis; photoreceptor cell maintenance; actin cytoskeleton organization and biogenesis; signal transduction; visual behavior Disease: Nephronophthisis 4; Senior-loken Syndrome 4
Research Articles on NPHP4	**1.** Study provides evidence that KIF13B and NPHP4 are both required for establishment of a specialized caveolin-1 membrane microdomain at the ciliary transition zone, which is essential for Shh-induced accumulation of SMO in the primary cilium as well as for activation of GLI-mediated target gene expression.
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