Nephrocystin-4 (NPHP4) ELISA Kit for Monkey

PRODUCT OVERVIEW

Product Name	Nephrocystin-4 (NPHP4), ELISA Kit
Full Product Name	Monkey Nephrocystin-4 (NPHP4) ELISA Kit
Product Gene Name	NPHP4 elisa kit
Species Reactivity	Monkey
Preparation and Storage	Store all reagents at 2-8 degree C
Molecular Weight	99,953 Da
OMIM	606966
Related Product Information for NPHP4 elisa kit	MBS9363393 is a ready-to-use microwell, strip plate ELISA (enzyme-linked immunosorbent assay) Kit for analyzing the presence of the Nephrocystin-4 (NPHP4) ELISA Kit target analytes in biological samples. The concentration gradients of the kit standards or positive controls render a theoretical kit detection range in biological research samples containing NPHP4. The ELISA analytical biochemical technique of the MBS9363393 kit is based on NPHP4 antibody-NPHP4 antigen interactions (immunosorbency) and an HRP colorimetric detection system to detect NPHP4 antigen targets in samples. The ELISA Kit is designed to detect native, not recombinant, NPHP4. Appropriate sample types may include undiluted body fluids and/or tissue homogenates, secretions. Quality control assays assessing reproducibility identified the intra-assay CV (%) and inter-assay CV(%).
Other Notes	Small volumes of NPHP4 elisa kit 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.

ADDITIONAL DETAILS

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

NCBI GI #	615276303
NCBI GeneID	261734
NCBI Accession #	NP_001278523.1
UniProt Secondary Accession #	Q8IWC0
UniProt Related Accession #	O75161
GenBank Nucleotide #	NM_001291594.1
NCBI Official Full Name	nephrocystin-4 isoform c
NCBI Official Synonym Full Names	nephronophthisis 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; cytosol; intercellular junction; membrane; photoreceptor connecting cilium; tight junction Molecular Function: protein binding; structural molecule activity Biological Process: actin cytoskeleton organization and biogenesis; cell-cell adhesion; organelle organization and biogenesis; photoreceptor cell maintenance; retina development in camera-type eye; signal transduction; sperm motility; visual behavior Disease: Nephronophthisis 4; Senior-loken Syndrome 4
Research Articles on NPHP4	**1.** homozygous NPHP4 truncating mutation that expands the phenotypic spectrum of NPHP4-related nephronophthisis to also include cerebello-oculo-renal syndrome and abnormal spermatogenesis causing male infertility
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