G-Biosciences Image not found or type unknown

Immunotag™ NPHP1 Polyclonal Antibody

Antibody Specification

Catalog No.	ITN0942
Product Description	Immunotag™ NPHP1 Polyclonal Antibody
Size	50 µg, 100 µg
Conjugation	HRP, Biotin, FITC, Alexa Fluor® 350, Alexa Fluor® 405, Alexa Fluor® 488, Alexa Fluor® 555, Alexa Fluor® 594, Alexa Fluor® 647
IMPORTANT NOTE	This product is custom manufactured with a lead time of 3-4 weeks. Once in production, this item cannot be cancelled from an order and is not eligible for return.
Target Protein	NPHP1
Clonality	Polyclonal
Storage/Stability	-20°C/1 year
Application	WB,ELISA
Recommended Dilution	WB 1:500-2000 ELISA 1:5000-20000
Concentration	1 mg/ml
Reactive Species	Human,Mouse
Host Species	Rabbit
Immunogen	Synthesized peptide derived from human protein . at AA range: 510-590
Specificity	NPHP1 Polyclonal Antibody detects endogenous levels of protein.
Purification	The antibody was affinity-purified from rabbit antiserum by affinity-chromatography using epitope-specific immunogen
Form	Liquid in PBS containing 50% glycerol, and 0.02% sodium azide.
Gene Name	NPHP1 NPH1
Accession No.	O15259 Q9QY53

Antibody Specification

Description	nephrocystin 1(NPHP1) Homo sapiens This gene encodes a protein with src homology domain 3 (SH3) patterns. This protein interacts with Crk-associated substrate, and it appears to function in the control of cell division, as well as in cell-cell and cell-matrix adhesion signaling, likely as part of a multifunctional complex localized in actin- and microtubule-based structures. Mutations in this gene cause familial juvenile nephronophthisis type 1, a kidney disorder involving both tubules and glomeruli. Defects in this gene are also associated with Senior-Loken syndrome type 1, also referred to as juvenile nephronophthisis with Leber amaurosis, which is characterized by kidney and eye disease, and with Joubert syndrome type 4, which is characterized by cerebellar ataxia, oculomotor apraxia, psychomotor delay and neonatal breathing abnormalities, sometimes including retinal dystrophy and renal disease. Multiple transcript variants encoding diffe
Protein Expression	Fetal brain,Fetal kidney,Human fetal kidney,
Subcellular Localization	cytosol,cytoskeleton,cell-cell junction,adherens junction,bicellular tight junction,cilium,membrane,motile cilium,photoreceptor connecting cilium,
Protein Function	disease:Defects in NPHP1 are the cause of Joubert syndrome type 4 (JBTS4) [MIM:609583]. JBTS is an autosomal recessive disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. JBTS4 is a phenotypically mild form.,disease:Defects in NPHP1 are the cause of nephronophthisis type 1 (NPHP1) [MIM:256100]; also known as familial juvenile nephronophthisis 1. NPHP1 is an autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years.,disease:Defects in NPHP1 are the cause of Senior-Loken syndrome type 1 (SLSN1) [MIM:266900]; also known as juvenile nephronophthisis with Leber amaurosis. 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.,domain:The SH3 domain mediates the stable interaction with Cas.,function:Together with Cas it may play a role in the control of epithelial cell polarity. Seems to help to recruit protein tyrosine kinase 2 beta (PTK2B) to cell matrix adhesions, thereby initiating phosphorylation of PTK2B and PTK2B-dependent signaling.,similarity:Belongs to the nephrocystin-1 family.,similarity:Contains 1 SH3 domain.,subcellular location:Localizes at or near the cell-cell adherens junctions.,subunit:Interacts with Crk-associated substrate (Cas), NPHP4, PTK2B and tensin. Interacts with INVS and NPHP3.,tissue specificity:Widespread expression, with highest levels in pituitary gland, spinal cord, thyroid gland, testis, skeletal muscle, lymph node and trachea. Weakly expressed in heart, kidney and pancreas.,
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