NPHP1 Monoclonal Antibody

Catalog No	BYmab-05815
Isotype	IgG
Reactivity	Human;Mouse
Applications	WB
Gene Name	NPHP1 NPH1
Protein Name	Nephrocystin-1 (Juvenile nephronophthisis 1 protein)
Immunogen	Synthesized peptide derived from human protein . at AA range: 510-590
Specificity	NPHP1 Monoclonal Antibody detects endogenous levels of protein.
Formulation	Liquid in PBS containing 50% glycerol, and 0.02% sodium azide.
Source	Monoclonal, Mouse,IgG
Purification	The antibody was affinity-purified from mouse antiserum by affinity-chromatography using epitope-specific immunogen.
Dilution	WB 1:500-2000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	
Observed Band	80kD
Cell Pathway	Cell junction . Cell junction, adherens junction . Cell projection, cilium . Cytoplasm, cytoskeleton, cilium axoneme . Cell junction, tight junction. In the retinal photoreceptor cell layer, localizes at the connecting cilium (By similarity). Colocalizes with E-cadherin and BCAR1 at or near the cell-cell adherens junctions (By similarity). Localized to respiratory cilia axoneme (PubMed:16308564, PubMed:16885411). Localized to the transition zone of respiratory cilia (PubMed:16885411). Localized to the transition zone of photoreceptor-connecting cilia and renal monocilia (By similarity). In cultured renal cells, it localizes diffusely in the cytoplasm but, as cells approach confluence, it accumulates at basolateral tight junctions (By similarity). .
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. Expressed in nasal epithelial cells (at protein level) (PubMed:16308564). Expressed in the renal collecting duct (at protein level) (PubMed:18477472).

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. Sy
Background	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
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