DNAxPAb | Hard-to-Find Antibody

NPHP1 DNAxPab

Catalog # H00004867-W01P Size 200 ug

Specification

Product Description	Rabbit polyclonal antibody raised against a full-length human NPHP1 DNA using DNAx™ Immune te chnology.
Technology	DNAx™ Immune
Immunogen	Full-length human DNA
Sequence	MLARRQRDPLQALRRRNQELKQQVDSLLSESQLKEALEPNKRQHIYQRCIQLKQAIDENKNALQK LSKADESAPVANYNQRKEEEHTLLDKLTQQLQGLAVTISRENITEYASFLPFFFLF
Host	Rabbit
Reactivity	Human
Purification	Protein A
Quality Control Testing	Antibody reactive against mammalian transfected lysate.
Storage Buffer	In 1x PBS, pH 7.4
Storage Instruction	Store at -20°C or lower. Aliquot to avoid repeated freezing and thawing.

Applications

- Western Blot (Transfected lysate)

  Protocol Download

- Immunofluorescence (Transfected cell)
- Flow Cytometry (Transfected cell)

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Gene Info — NPHP1

Entrez GeneID	4867
GeneBank Accession#	NM_207181.1
Protein Accession#	NP_997064.1
Gene Name	NPHP1
Gene Alias	FLJ97602, JBTS4, NPH1, SLSN1
Gene Description	nephronophthisis 1 (juvenile)
Omim ID	256100 266900 607100 609583
Gene Ontology	Hyperlink
Gene Summary	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 different isoforms have been found for this gene. [provided by RefSeq
Other Designations	nephrocystin-1

Disease

- Abnormalities
- Cerebellar Ataxia
- Genetic Predisposition to Disease
- Hyperparathyroidism
- Kidney Diseases
- Mental Retardation
- Syndrome

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