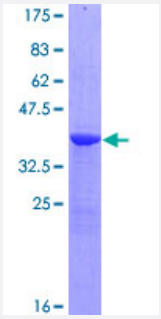
Full-Length

NPHP1 (Human) Recombinant Protein (P01)

Catalog # H00004867-P01 Size 25 ug, 10 ug

Applications



Specification

Product Description	Human NPHP1 full-length ORF (NP_997064.1, 1 a.a. - 121 a.a.) recombinant protein with GST-tag at N-terminal.
Sequence	MLARRQRDPLQALRRRNQELKQQVDSLLSESQLKEALEPNKRQHIYQRCIQLKQAIDENKNALQK LSKADESAPVANYNQRKEEEHTLLDKLTQQLQGLAVTISRENITEYASFLPFFFLF
Host	Wheat Germ (in vitro)
Theoretical MW (kDa)	40.7
Preparation Method	*in vitro* wheat germ expression system
Purification	Glutathione Sepharose 4 Fast Flow
Quality Control Testing	12.5% SDS-PAGE Stained with Coomassie Blue.
Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.
Storage Instruction	Store at -80°C. Aliquot to avoid repeated freezing and thawing.
Note	Best use within three months from the date of receipt of this protein.

Applications

- Enzyme-linked Immunoabsorbent Assay
- Western Blot (Recombinant protein)
- Antibody Production
- Protein Array

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