

## NPHP1 mouse monoclonal antibody (hybridoma)

Catalog # H00004867-M Size Up to 5 Clones

Specification	
Product Description	Mouse monoclonal antibody raised against a full-length recombinant NPHP1.
lmmunogen	NPHP1 (NP_997064.1, 1 a.a. ~ 121 a.a) full-length recombinant protein with GST tag. MW of the GS T tag alone is 26 KDa.
Sequence	MLARRQRDPLQALRRRNQELKQQVDSLLSESQLKEALEPNKRQHIYQRCIQLKQAIDENKNALQK LSKADESAPVANYNQRKEEEHTLLDKLTQQLQGLAVTISRENITEYASFLPFFFLF
Host	Mouse
Reactivity	Human
Quality Control Testing	Antibody reactivity and specificity confirmed by ELISA and Western Blot.
Deliverables	Up to 5 positive hybridoma clones will be delivered to customer in the cryotube format.
Note	Customer should check the viability of the hybridomas within one month from the date of receipt. Fee -for-service of long term hybridoma storage can be performed upon customer's request.

## Applications

Western Blot (Transfected lysate)

Protocol Download

Western Blot (Recombinant protein)

**Protocol Download** 

ELISA

## Gene Info — NPHP1



Entrez GeneID	<u>4867</u>
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	<u>256100</u> <u>266900</u> <u>607100</u> <u>609583</u>
Gene Ontology	<u>Hyperlink</u>
Gene Summary	This gene encodes a protein with src homology domain 3 (SH3) patterns. This protein interacts wi th Crk-associated substrate, and it appears to function in the control of cell division, as well as in c ell-cell and cell-matrix adhesion signaling, likely as part of a multifunctional complex localized in ac tin- and microtubule-based structures. Mutations in this gene cause familial juvenile nephronophthi sis type 1, a kidney disorder involving both tubules and glomeruli. Defects in this gene are also as sociated with Senior-Loken syndrome type 1, also referred to as juvenile nephronophthisis with L eber 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 neo natal breathing abnormalities, sometimes including retinal dystrophy and renal disease. Multiple tr anscript 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