Recombinant Human NPHP1 cell lysate

Cat. No. NPHP1-1210HCL **Lot. No.** (See product label)

SPECIFICATION

Product Overview	
Species	Human
Description	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.
Size	100 ul
Storage Buffer	1X Sample Buffer (50 mM Tris-HCl, 2% SDS, 10% glycerol, 300 mM 2-mercaptoethanol, 0.01% Bromophenol blue)
Applications	Western Blot;

GENE INFORMATION

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Gene Name	NPHP1 nephronophthisis 1 (juvenile) [Homo sapiens]
Official Symbol	NPHP1
Synonyms	NPHP1; nephronophthisis 1 (juvenile); NPH1; nephrocystin-1; JBTS4; nephrocystin 1; juvenile nephronophthisis 1 protein; SLSN1; FLJ97602;
Gene ID	4867
mRNA Refseq	NM_000272
Protein Refseq	NP_000263
MIM	607100
UniProt ID	O15259
Chromosome Location	2q13
Function	protein binding; structural molecule activity;

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