Recombinant Full Length Human NPHP1 Protein, GST-tagged

Cat. No. NPHP1-6620HF **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human NPHP1 full-length ORF (NP_997064.1, 1 a.a. - 121 a.a.) recombinant protein with GST-tag at N-terminal.
Species	Human
Source	In Vitro Cell Free System
ProteinLength	121 amino acids
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. [provided by RefSeq
Molecular Mass	40.7 kDa
AA Sequence	MLARRQRDPLQALRRRNQELKQQVDSLLSESQLKEALEPNKRQHIYQRCIQLKQAID

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	ENKNALQKLSKADESAPVANYNQRKEEEHTLLDKLTQQLQGLAVTISRENITEYASFL PFFFLF
Applications	Enzyme-linked Immunoabsorbent Assay Western Blot (Recombinant protein) Antibody Production Protein Array
Notes	Best use within three months from the date of receipt of this protein.
Storage	Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.
Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

GENE INFORMATION

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

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UniProt ID	O15259

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