

## Recombinant Human NPHP1 Protein, GST-tagged

**Cat. No.** NPHP1-6026H    **Lot. No.** (See product label)

### SPECIFICATION

|                         |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
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| <b>Product Overview</b> | Human NPHP1 full-length ORF ( NP_997064.1, 1 a.a. - 121 a.a.) recombinant protein with GST-tag at N-terminal.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          |
| <b>Species</b>          | Human                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |
| <b>Source</b>           | Wheat Germ                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
| <b>Description</b>      | <p>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.</p> <p>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</p> |
| <b>Molecular Mass</b>   | 40.7 kDa                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               |
| <b>AA Sequence</b>      | MLARRQRDPLQALRRRNQELKQQVDSLLSESQLKEALEPNKRQHYYQRCIQLKQAID<br>ENKNALQKLSKADESAPVANYNQRKEEEHTLLDKLTQQLQGLAVTISRENITEYASFL                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |

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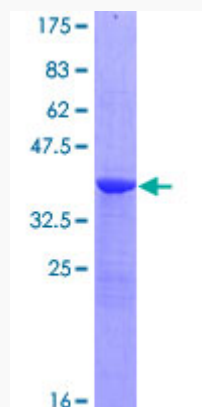
 45-1 Ramsey Road, Shirley, NY 11967, USA

|                         |                                                                                                                                          |
|-------------------------|------------------------------------------------------------------------------------------------------------------------------------------|
|                         | PFFFLF                                                                                                                                   |
| <b>Applications</b>     | Enzyme-linked Immunoabsorbent Assay<br>Western Blot (Recombinant protein)<br>Antibody Production<br>Protein Array                        |
| <b>Notes</b>            | Best use within three months from the date of receipt of this protein.                                                                   |
| <b>Storage</b>          | Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.                                                                 |
| <b>Storage Buffer</b>   | 50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.                                                                 |
| <b>GENE INFORMATION</b> |                                                                                                                                          |
| <b>Gene Name</b>        | NPHP1 nephronophthisis 1 (juvenile) [ Homo sapiens ]                                                                                     |
| <b>Official Symbol</b>  | NPHP1                                                                                                                                    |
| <b>Synonyms</b>         | NPHP1; nephronophthisis 1 (juvenile); NPH1; nephrocystin-1; JBTS4; nephrocystin 1; juvenile nephronophthisis 1 protein; SLSN1; FLJ97602; |
| <b>Gene ID</b>          | 4867                                                                                                                                     |
| <b>mRNA Refseq</b>      | NM_000272                                                                                                                                |
| <b>Protein Refseq</b>   | NP_000263                                                                                                                                |
| <b>MIM</b>              | 607100                                                                                                                                   |
| <b>UniProt ID</b>       | O15259                                                                                                                                   |

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**Quality Control****Testing:**

12.5% SDS-PAGE Stained with Coomassie Blue.

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