

## NPHP1

**Reactivity:** Human Mouse Rat

**Tested applications:** WB IHC IF

**Recommended Dilution:** WB 1:500 - 1:2000 IHC 1:50 - 1:200 IF 1:50 - 1:100

**Calculated MW:** 83kDa

**Observed MW:** Refer to Figures

**Immunogen:**

Recombinant protein of human NPHP1

**Storage Buffer:**

Store at -20. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

**Synonym:**

NPH1; JBTS4; SLSN1;

**Catalog #:** A6674

**Antibody Type:**

Polyclonal Antibody

**Species:** Rabbit

**Gene ID:** 4867

**Isotype:** IgG

**Swiss Prot:** O15259

**Purity:** Affinity purification

For research use only.

**Background:**

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.

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