Recombinant Human NPHP1

Catalog No: #GP13468

Package Size: #GP13468-1 100ug

Orders: order@signalwayantibody.com
Support: tech@signalwayantibody.com

Description

Product Name	Recombinant Human NPHP1
Brief Description	Recombinant Protein
Immunogen Description	Fusion protein corresponding to a region derived from 1-110 amino acids of human NPHP1
Target Name	nephrocystin 1
Other Names	NPH1; JBTS4; SLSN1
Accession No.	Swissprot:O15259Gene Accession:BC009789
Uniprot	O15259
GeneID	4867;
Storage	-20~-80°C, pH 7.6 PBS

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.

References

Note: For in vitro research use only, not for diagnostic or therapeutic use. This product is not a medical device.

Note: This product is for in vitro research use only