NPHP1 Polyclonal Antibody

Cat No: HR1AP10999

For research use only

Overview

Product Name	NPHP1 Polyclonal Antibody
Source	Rabbit
Applications	WB,ELISA
Species Reactivity	Human,Mouse
Recommended Dilutions	
Immunogen	
Species	Rabbit
Storage	-20°C/1 year
Isotype	
Clonality	
Concentration	1 mg/ml
Observed band	80kDa
GeneID?Human?	NPHP1 NPH1
Human Swiss-Prot No.	
Cellular localization	
Alternative Names	
Background	nephrocystin 1(NPHP1) Homo sapiens 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 diffe