

Product name:	Nephrocystin-4 Rabbit Polyclonal Antibody
Cat number:	ABN14563
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human NPHP4. AA range:877-926
Reactivity:	Human,Rat,Mouse
Applications:	IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:20000-1:40000
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

This gene encodes a protein involved in renal tubular development and function. This protein interacts with nephrocystin, and belongs to a multifunctional complex that is localized to actin- and microtubule-based structures. Mutations in this gene are associated with nephronophthisis type 4, a renal disease, and with Senior-Loken syndrome type 4, a combination of nephronophthisis and retinitis pigmentosa. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Apr 2014],disease:Defects in NPHP4 are the cause of nephronophthisis type 4 (NPHP4) [MIM:606966]; also known as familial juvenile nephronophthisis 4. NPHP4 is an autosomal recessive inherited disease resulting in end-stage renal disease at age ranging between 6 and 35 years. It is a progressive tubulo-interstitial kidney disorder characterized by polydipsia, polyuria, anemia and growth retardation. The most prominent histological features are modifications of the tubules with thickening of the basement membrane, interstitial fibrosis and, in the advanced stages, medullary cysts.,disease:Defects in NPHP4 are the cause of Senior-Loken syndrome type 4 (SLSN4) [MIM:606996]. SLSN is a renal-retinal disorder characterized by progressive wasting of the filtering unit of the kidney, with or without medullary cystic renal disease, and progressive eye disease. Typically this disorder becomes apparent during the first year of life.,similarity:Belongs to the NPHP4 family.,subunit:Interacts with NPHP1 and RPGRIP1L.,tissue specificity:Expressed in kidney, skeletal muscle, heart and liver, and to a lesser extent in brain and lung.,