

NPHP3 rabbit pAb

Catalog No :	YT6689
Reactivity :	Human;Mouse
Applications :	WB
Target :	NPHP3
Gene Name :	NPHP3 KIAA2000
Protein Name :	NPHP3
Human Gene Id :	27031
Human Swiss Prot No :	Q7Z494
Mouse Gene Id :	74025
Mouse Swiss Prot No :	Q7TNH6
Immunogen :	Synthesized peptide derived from human NPHP3 AA range: 1092-1142
Specificity :	This antibody detects endogenous levels of NPHP3 at Human/Mouse
Formulation :	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.
Source :	Polyclonal, Rabbit,IgG
Dilution :	WB 1:500-2000
Purification :	The antibody was affinity-purified from rabbit antiserum by affinity-chromatography using epitope-specific immunogen.
Concentration :	1 mg/ml
Storage Stability :	-15°C to -25°C/1 year(Do not lower than -25°C)

Molecularweight : 146kD**Background :**

This gene encodes a protein containing a coiled-coil (CC) domain, a tubulin-tyrosine ligase (TTL) domain, and a tetratricopeptide repeat (TPR) domain. The encoded protein interacts with nephrocystin, it is required for normal ciliary development, and it functions in renal tubular development. Mutations in this gene are associated with nephronophthisis type 3, and also with renal-hepatic-pancreatic dysplasia, and Meckel syndrome type 7. Naturally occurring read-through transcripts exist between this gene and the downstream ACAD11 (acyl-CoA dehydrogenase family, member 11) gene. [provided by RefSeq, Feb 2011],

Function :

alternative products:Additional isoforms seem to exist,disease:Defects in NPHP3 are a cause of renal-hepatic-pancreatic dysplasia (RHPD) [MIM:208540]. RHPD is an autosomal recessive disorder with variable expression, and patients surviving the neonatal period progress to renal and hepatic failure which can be treated successfully with combined liver-kidney transplantation.,disease:Defects in NPHP3 are the cause of nephronophthisis type 3 (NPHP3) [MIM:604387]; also known as adolescent nephronophthisis. NPHP3 is a autosomal recessive disorder resulting in end-stage renal disease. It is characterized by polyuria, polydipsia, anemia. Onset of terminal renal failure occurs significantly later (median age, 19 years) than in juvenile nephronophthisis. Renal pathology is characterized by alterations of tubular basement membranes, tubular atrophy and dilatation, sclerosing tubulointerstitial neph

Subcellular Location :

Cell projection, cilium . Localization to cilium is mediated via interaction with UNC119 and UNC119B, which bind to the myristoyl moiety of the N-terminus.

Expression :

Widely expressed at low level. Expressed in heart, placenta, liver, skeletal muscle, kidney and pancreas. Expressed at very low level in brain and lung.

Products Images

