NPHP3 蛋白抗体

产品货号： mlR19329

英文名称： NPHP3

中文名称： NPHP3 蛋白抗体

别　　名： Meckel syndrome, type 7; MKS7; Nephrocystin-3; nephronophthisis 3 (adolescent); NPH3; Nphp3; NPHP3_HUMAN; pcy; RHPD.

研究领域： 细胞生物　发育生物学　信号转导

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Pig, Cow, Horse, Sheep,

产品应用： ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500 （石蜡切片需做抗原修复）
not yet tested in other applications.
optimal dilutions/concentrations should be determined by the end user.

分 子 量： 150kDa

细胞定位： 细胞核

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human NPHP3:131-230/1330

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： Store at -20 ° C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20° C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 ° C.

PubMed： PubMed

产品介绍： This gene encodes a protein containing a coiled-coil (CC) domain, a tubulin-tyrosine ligase (TTL) domain, and a tetratrico peptide 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:

Required for normal ciliary development and function. Inhibits disheveled-1-induced canonical Wnt-signaling activity and may also play a role in the control of non-canonical Wnt signaling which regulates planar cell polarity. Probably acts as a molecular switch between different Wnt signaling pathways. Required for proper convergent extension cell movements.

Subcellular Location:

Cell projection; cilium.

Tissue Specificity:

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

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 occurr significantly later (median age, 19 years) than in juvenile nephronophthisis. Renal pathology is characterized by alterations of tubular basement membranes, tubular atrophy and dilation, sclerosing tubulointerstitial nephropathy, and renal cyst development predominantly at the corticomedullary junction. 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.

Defects in NPHP3 are the cause of Meckel syndrome type 7 (MKS7) [MIM:267010]. It is a form of Meckel syndrome, an autosomal recessive disorder. It is characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly.

Similarity:

Contains 11 TPR repeats.

SWISS:

Q7Z494

Gene ID:

27031

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.