abcam

Product datasheet

Recombinant Human NPHP3 protein ab162057

1 Image

Overview

Product name	Recombinant Human NPHP3 protein
Protein length	Protein fragment

Description

Nature	Recombinant
Source	Wheat germ
Amino Acid Sequence	
Species	Human
Sequence	NQELLSMGRREAKLDTENKRLRAELQALQKTYQKILRE KESALEAKYQAM ERAATFEHDRDKVKRQFKIFRETKENEIQDLLRAKREL ESKLQRLQAQGI
Amino acids	106 to 205
Tags	GST tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab162057** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	Western blot ELISA
Form	Liquid
Additional notes	Protein concentration is above or equal to 0.05 mg/ml.

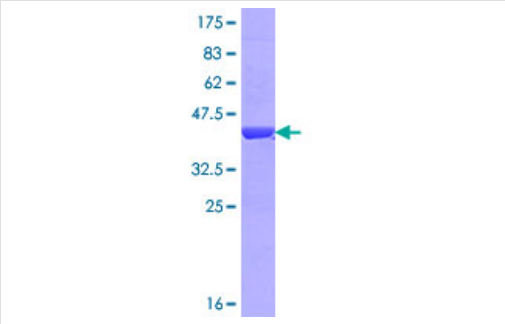
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

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.
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.
Involvement in 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.
Sequence similarities	Contains 11 TPR repeats.
Cellular localization	Cell projection > cilium.

Images



SDS-PAGE - Recombinant Human NPHP3 protein (ab162057)

ab162057 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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