



# MKKS rabbit pAb

Catalog No	YP-Ab-08069
Isotype	IgG
Reactivity	Human; Mouse
Applications	WB
Gene Name	MKKS BBS6
Protein Name	MKKS
Immunogen	Synthesized peptide derived from human MKKS AA range: 166-216
Specificity	This antibody detects endogenous levels of MKKS at Human/Mouse
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.184% sodium azide.
Source	Polyclonal, Rabbit,IgG
Purification	The antibody was affinity-purified from rabbit serum by affinity-chromatography using specific immunogen.
Dilution	WB 1:500-2000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	McKusick-Kaufman/Bardet-Biedl syndromes putative chaperonin (Bardet-Biedl syndrome 6 protein)
Observed Band	65kD
Cell Pathway	Cytoplasm, cytoskeleton, microtubule organizing center, centrosome. Cytoplasm, cytosol . Nucleus . The majority of the protein resides within the pericentriolar material (PCM), a proteinaceous tube surrounding centrioles. During interphase, the protein is confined to the lateral surfaces of the PCM but during mitosis it relocalizes throughout the PCM and is found at the intercellular bridge. The MKSS protein is highly mobile and rapidly shuttles between the cytosol and centrosome.
Tissue Specificity	Widely expressed in adult and fetal tissues.
Function	disease:Defects in MKKS are the cause of Bardet-Biedl syndrome type 6 (BBS6) [MIM:209900]. Bardet-Biedl syndrome (BBS) is a genetically heterogeneous, autosomal recessive disorder characterized by usually severe pigmentary retinopathy, early onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. A relatively high incidence of BBS is found in the mixed Arab populations of Kuwait and in Bedouin tribes throughout the Middle East, most likely due to the high rate of consaguinity in these populations and a founder effect, disease:Defects in MKKS are the cause of McKusick-Kaufman syndrome (MKKS) [MIM:236700]. MKKS is an autosomal recessive developmental disorder. It is characterized by hydrometrocolpos,



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postaxial polydactyly and congenital heart defects.,function:May play a

### Background

This gene encodes a protein which shares sequence similarity with other members of the type II chaperonin family. The encoded protein is a centrosome-shuttling protein and plays an important role in cytokinesis. This protein also interacts with other type II chaperonin members to form a complex known as the BBSome, which involves ciliary membrane biogenesis. This protein is encoded by a downstream open reading frame (dORF). Several upstream open reading frames (uORFs) have been identified, which repress the translation of the dORF, and two of which can encode small mitochondrial membrane proteins. Mutations in this gene have been observed in patients with Bardet-Biedl syndrome type 6, also known as McKusick-Kaufman syndrome. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Nov 2013],

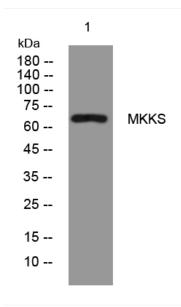
## matters needing attention

Avoid repeated freezing and thawing!

#### **Usage suggestions**

This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.

### **Products Images**



Western blot analysis of lysates from HuvEc cells, primary antibody was diluted at 1:1000, 4° over night