驱动蛋白家族蛋白 7 抗体

产品货号： mlR12388

英文名称： KIF7

中文名称： 驱动蛋白家族蛋白 7 抗体

别　　名： EQYK340; kif 7; Kif-7; KIF7_HUMAN; kinesin family member 7; kinesin like protein KIF7; Kinesin-like protein kif7; UNQ340.

研究领域： 细胞生物　干细胞　细胞骨架

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Dog,

产品应用： WB=1:500-2000 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： 细胞浆

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human KIF7:751-850/1343

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： KIF7 is a 1,343 amino acid protein expressed in embryonic stem cells, melanotic melanoma and Jurkat T-cells. KIF7 is a member of the KIF27 subfamily of the kinesin-like protein family and contains one kinesin-motor domain. It is suggested that KIF7 may participate in the Hedgehog (Hh) signaling pathway by regulating the proteolysis and stability of GLI transcription factors. Hedgehog (Hh) signaling plays a critical role in embryonic development.

Function:

Acts as both a negative and positive regulator of sonic hedgehog (Shh) pathway, acting downstream of SMO. Negatively regulates the pathway by preventing inappropriate activation of the transcriptional activator GLI2 in the absence of ligand. Positively regulates the pathway by preventing the processing of the transcription factor GLI3 into its repressor form. Required for efficient localization of GLI3 to cilia in response to Shh. May also act as a ciliary motor.

Subunit:

Interacts with GLI1, GLI2, GLI3, SMO and SUFU. Interacts with NPHP1.

Subcellular Location:

Cell projection; cilium. SMO is required for its accumulation within cilia. Moves from the cilia base to the cilia tip in response to activation of the Shh pathway.

Tissue Specificity:

Embryonic stem cells, melanotic melanoma and Jurkat T-cells.

DISEASE:

Note=Ciliary dysfunction leads to a broad spectrum of disorders, collectively termed ciliopathies. The ciliopathy range of diseases includes Meckel-Gruber syndrome, Bardet-Biedl syndrome, Joubert syndrome, and hydrolethalus syndrome among others. Single-locus allelism is insufficient to explain the variable penetrance and expressivity of such disorders, leading to the suggestion that variations across multiple sites of the ciliary proteome influence the clinical outcome. Primary ciliopathy loci can be modulated by pathogenic lesions in other ciliary genes to either exacerbate overall severity or induce specific endophenotypes. KIF7 may be causally associated with diverse ciliopathies, and also acts as a modifier gene across the ciliopathy spectrum.

Defects in KIF7 may be a cause of Bardet-Biedl syndrome (BBS) [MIM:209900]. A syndrome 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. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. Note=Heterozygous missense mutations in KIF7 may genetically interact with other BBS genes and contribute to disease manifestation and severity.

Defects in KIF7 are the cause of hydrolethalus syndrome type 2 (HLS2) [MIM:614120]. HLS2 is an embryonic lethal disorder characterized by hydrocephaly or anencephaly, postaxial polydactyly of the upper limbs, and pre- or postaxial polydactyly of the lower limbs. Duplication of the hallux is a common finding.

Defects in KIF7 are the cause of acrocallosal syndrome (ACLS) [MIM:200990]. ACLS is a syndrome that is characterized by postaxial polydactyly, hallux duplication, macrocephaly and absence of the corpus callosum, usually with severe developmental delay.

Defects in KIF7 are the cause of Joubert syndrome type 12 (JBTS12) [MIM:200990]. JBTS12 is a disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease.

Defects in KIF7 may be a cause of Pallister-Hall syndrome (PHS) [MIM:146510]. An autosomal dominant disorder

characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia.

Similarity:

Belongs to the kinesin-like protein family. KIF27 subfamily.

Contains 1 kinesin-motor domain.

SWISS:

Q2M1P5

Gene ID:

374654

Important Note:

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

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