Kartagener 综合征相关蛋白 RSHL3 抗体

产品货号： mlR11472

英文名称： RSPH4A

中文名称： Kartagener 综合征相关蛋白 RSHL3 抗体

别　　名： CILD11; dJ412I7.1; Radial spoke head protein 4 homolog A; Radial spoke head-like protein 3; RSH4A_HUMAN; RSHL3; Rsph4a; RSPH6B; A230081C05.

研究领域： 细胞生物　发育生物学　神经生物学　信号转导　细胞骨架　细胞外基质

抗体来源： Rabbit

克隆类型： Polyclonal

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

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： 81kDa

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 RSPH4A/RSHL3:435-482/716

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： RSHL3 is predicted to be a component of the radial spoke head based on homology with proteins in the biflagellate alga Chlamydomonas reinhardtii and other ciliates. RSHL3 (radial spoke head-like protein 3), also known as radial spoke head protein 4 homolog A, is a 716 amino acid protein that belongs to the flagellar

radial spoke RSP4/6 family. Mutations in the RSHL3 gene cause primary ciliary dyskinesia 1, a disease arising from dysmotility of motile cilia and sperm. Existing as three alternatively spliced isoforms, the RSHL3 gene contains 6 exons, is conserved in chimpanzee, dog, cow, mouse, rat, chicken, zebrafish, fruit fly and P.falciparum, and maps to human chromosome 6q22.1.

Function:

Probable component of the axonemal radial spoke head. Radial spokes are regularly spaced along cilia, sperm and flagella axonemes. They consist of a thin stalk which is attached to a subfiber of the outer doublet microtubule, and a bulbous head which is attached to the stalk and appears to interact with the projections from the central pair of microtubules.

Subcellular Location:

Cytoplasm; cytoskeleton; cilium axoneme. Radial spoke.

Tissue Specificity:

Defects in RSPH4A are the cause of primary ciliary dyskinesia type 11 (CILD11) [MIM:612649]. CILD is an autosomal recessive disorder characterized by axonemal abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit situs inversus, due to dysfunction of monocilia at the embryonic node and randomization of left-right body asymmetry. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome.

DISEASE:

Defects in RSPH4A are the cause of primary ciliary dyskinesia type 11 (CILD11) [MIM:612649]. CILD is an autosomal recessive disorder characterized by axonemal abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit situs inversus, due to dysfunction of monocilia at the embryonic node and randomization of left-right body asymmetry. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome.

Similarity:

Belongs to the flagellar radial spoke RSP4/6 family.

SWISS:

Q5TD94

Gene ID:

345895

Important Note:

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

Kartagener 综合征:由下列三联症组成，支气管扩张、鼻窦炎或鼻息肉及内脏反位(主要是右位心）。若仅具备内脏反位及支气管扩张两项，则为不全性 Kartagener 综合征。常合并其他先天性畸形。其病因是由于全身纤毛先天性缺乏轴丝臂，引起纤毛活动力丧失、黏液纤毛运输功能障碍，分泌物和细菌潴留而发生持续性感染长期存在所致。以学龄儿童及青少年多发，有家族史。主要症状为随年龄加重的咳嗽、咳痰和咯血，晨起明显，易患感冒及肺炎，常见体征为发绀和杵状指。

产品图片