诺里病 NDP 蛋白/早产儿视网膜病蛋白抗体

产品货号： mlR19062

英文名称： NORRIN

中文名称： 诺里病 NDP 蛋白/早产儿视网膜病蛋白抗体

别　　名： EVR2; Exudative vitreoretinopathy 2 (X linked); FEVR; ND; NDP; NDP_HUMAN; Norrie disease (pseudoglioma); Norrie disease protein; Norrin; Norrin precursor; X linked exudative vitreoretinopathy 2 protein; X-linked exudative vitreoretinopathy 2 protein.

研究领域： 细胞生物　发育生物学　神经生物学　干细胞

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Dog, Pig, Cow, Horse, Rabbit, 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： 12kDa

细胞定位： 分泌型蛋白

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human NORRIN:51-133/133

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 secreted protein with a cystein-knot motif that activates the Wnt/beta-catenin pathway. The protein forms disulfide-linked oligomers in the extracellular matrix. Mutations in this gene result in Norrie disease and X-linked exudative vitreoretinopathy. [provided by RefSeq, Feb 2009]

Function:

Activates the canonical Wnt signaling pathway through FZD4 and LRP5 coreceptor. Plays a central role in retinal vascularization by acting as a ligand for FZD4 that signals via stabilizing beta-catenin (CTNNB1) and activating LEF/TCF-mediated transcriptional programs. Acts in concert with TSPAN12 to activate FZD4 independently of the Wnt-dependent activation of FZD4, suggesting the existence of a Wnt-independent signaling that also promote accumulation the beta-catenin (CTNNB1). May be involved in a pathway that regulates neural cell differentiation and proliferation. Possible role in neuroectodermal cell-cell interaction.

Subcellular Location:

Secreted.

Tissue Specificity:

Expressed in the outer nuclear, inner nuclear and ganglion cell layers of the retina, and in fetal and adult brain.

DISEASE:

Defects in NDP are the cause of Norrie disease (ND) [MIM:310600]; also known as atrophia bulborum hereditaria or Episkopi blindness. ND is a recessive disorder characterized by very early childhood blindness due to degenerative and proliferative changes of the neuroretina. Approximately 50% of patients show some form of progressive mental disorder, often with psychotic features, and about one-third of patients develop sensorineural deafness in the second decade. In addition, some patients have more complex phenotypes, including growth failure and seizure. Defects in NDP are the cause of vitreoretinopathy exudative type 2 (EVR2) [MIM:305390]. EVR2 is a disorder of the retinal vasculature characterized by an abrupt cessation of growth of peripheral capillaries, leading to an avascular peripheral retina. This may lead to compensatory retinal neovascularization, which is thought to be induced by hypoxia from the initial avascular insult. New vessels are prone to leakage and rupture causing exudates and bleeding, followed by scarring, retinal detachment and blindness. Clinical features can be highly variable, even within the same family. Patients with mild forms of the disease are asymptomatic, and their only disease related abnormality is an arc of avascular retina in the extreme temporal periphery.

Similarity:

Contains 1 CTCK (C-terminal cystine knot-like) domain.

SWISS:

Q00604

Gene ID:

4693

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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