

Product name:	MFRP Rabbit Polyclonal Antibody
Cat number:	ABN13851
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	Synthesized peptide derived from human MFRP
Reactivity:	Human,Rat,Mouse
Applications:	WB 1:500-1:2000,IHC 1:50-1:200,ICC/IF 1:50-1:200
Molecular Weight:	62kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

membrane frizzled-related protein(MFRP) Homo sapiens This gene encodes a member of the frizzled-related protein family. The encoded protein plays an important role in eye development and mutations in this gene have been associated with nanophthalmos, posterior microphthalmia, retinitis pigmentosa, foveoschisis, and optic disc drusen. The protein is encoded by a bicistronic transcript which also encodes C1q and tumor necrosis factor related protein 5 (C1QTNF5). [provided by RefSeq, Jun 2013],developmental stage:Expressed in fetal brain.,disease:Defects in C1QTNF5 are a cause of late-onset retinal degeneration (LORD) [MIM:605670]. LORD is an autosomal dominant disorder characterized by onset in the fifth to sixth decade with night blindness and punctate yellow-white deposits in the retinal fundus, progressing to severe central and peripheral degeneration, with choroidal neovascularization and chorioretinal atrophy.,disease:Defects in MFRP are the cause of microphthalmia MFRP-related (MCOPMFRP) [MIM:611040]. Microphthalmia is a clinically heterogeneous disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues. Ocular abnormalities like opacities of the cornea and lens, scarring of the retina and choroid, cataract and other abnormalities like cataract may also be present. MCOPMFRP is characterized by posterior microphthalmia, retinitis pigmentosa, foveoschisis and optic disc drusen.,disease:Defects in MFRP are the cause of nanophthalmos 2 (NNO2) [MIM:609549]. NNO2 is a rare autosomal recessive disorder of eye development characterized by extreme hyperopia and small functional eyes.,function:May play a role in eye development.,similarity:Contains 1 C1q domain.,similarity:Contains 1 collagen-like domain.,similarity:Contains 1 FZ (frizzled) domain.,similarity:Contains 2 CUB domains.,similarity:Contains 2 LDL-receptor class A domains.,tissue specificity:Specifically expressed in brain. Strongly expressed in medulla oblongata and to a lower extent in hippocampus and corpus callosum. Expressed in keratinocytes.,