

CHM Antibody (Center)

Catalog_no :	AB2017
Reactivity :	H
Category :	抗原抗体
Size :	100μL/50μL
Immunogen :	HUMAN:292-320
Specificity :	This CHM antibody is generated from rabbits immunized with a KLH conjugated synthetic peptide between 292-320 amino acids from the Central region of human CHM.
Dilution :	WB,1:1000;
Purification :	Purified polyclonal antibody supplied in PBS with 0.09% (W/V) sodium azide. This antibody is purified through a protein A column, followed by peptide affinity purification.
Other_name :	Rab proteins geranylgeranyltransferase component A 1, Choroideremia protein, Rab escort protein 1, REP-1, TCD protein, CHM, REP1, TCD
Isotype :	Rabbit Ig
Background :	This gene encodes component A of the RAB geranylgeranyl transferase holoenzyme. In the dimeric holoenzyme, this subunit binds unprenylated Rab GTPases and then presents them to the catalytic Rab GGTase subunit for the geranylgeranyl transfer reaction. Rab GTPases need to be geranylgeranylated on either one or two cysteine residues in their C-terminus to localize to the correct intracellular membrane. Mutations in this gene are a cause of choroideremia; also known as tapetochoroidal dystrophy (TCD). This X-linked disease is characterized by progressive dystrophy of the choroid, retinal pigment epithelium and retina. Alternative splicing results in multiple transcript variants encoding different isoforms.
reference :	Perez-Cano, H.J., et al. Am. J. Med. Genet. A 149A (10), 2134-2140 (2009) ; Renner, A.B., et al. Arch. Ophthalmol. 127(7):907-912(2009) Sergeev, Y.V., et al. Mutat. Res. 665 (1-2), 44-50 (2009) ; MacDonald, I.M., et al. Surv Ophthalmol 54(3):401-407(