

CHM

Reactivity: Human

Tested applications: WB

Recommended Dilution: WB 1:500 - 1:2000

Calculated MW: 73kDa

Observed MW: Refer to figures

Immunogen:

Recombinant protein of human CHM

Storage Buffer:

Store at -20. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

Synonym:

TCD; GGTA; REP-1; DXS540; HSD-32;

Catalog #: A8345

Antibody Type:

Polyclonal Antibody

Species: Rabbit

Gene ID: 1121

Isotype: IgG

Swiss Prot: P24386

Purity: Affinity purification

For research use only.

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.

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