
ETHE1 Rabbit pAb

Catalog Number: bs-7610R

Target Protein: ETHE1

Concentration: 1mg/ml

Form: Liquid

Host: Rabbit

Clonality: Polyclonal

Isotype: IgG

Applications: ELISA (1:5000-10000)

Reactivity: (predicted:Human, Mouse, Rabbit, Pig, Sheep, Cow, Dog)

Predicted MW: 26 kDa

Entrez Gene: 23474

Swiss Prot: O95571

Source: KLH conjugated synthetic peptide derived from human ETHE1: 191-254/254.

Purification: affinity purified by Protein A

Storage: 0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.

Shipped at 4°C. Store at -20°C for one year. Avoid repeated freeze/thaw cycles.

Background: Probably plays an important role in metabolic homeostasis in mitochondria. May function as a nuclear-cytoplasmic shuttling protein that binds transcription factor RELA/NFKB3 in the nucleus and exports it to the cytoplasm. Suppresses p53-induced apoptosis by preventing nuclear localization of RELA.

Involvement in disease: Defects in ETHE1 are a cause of ethylmalonic encephalopathy (EE) . EE is an autosomal recessive disorder characterized by neurodevelopmental delay and regression, recurrent petechiae, acrocyanosis, diarrhea, leading to death in the first decade of life. It is also associated with persistent lactic acidemia and ethylmalonic and methylsuccinic aciduria.