

# TBC1D24 (G-6): sc-390237

## BACKGROUND

TBC1D24 (TBC1 domain family member 24) is a 559 amino acid cytoplasmic protein that may act as a GTPase-activating protein for Rab family proteins and exists as two alternatively spliced isoforms. TBC1D24 contains one Rab-GAP TBC domain, one TLD domain and interacts with ARF6. Involved in neuronal projection development, probably through a negative modulation of ARF6 function, TBC1D24 is highly expressed in brain. TBC1D24 is also expressed in testis, skeletal muscle, heart, kidney, lung and liver. Defects in the TBC1D24 gene are the cause of familial infantile myoclonic epilepsy (FIME), which is characterized as a subtype of idiopathic epilepsy starting in early infancy and manifesting as myoclonic seizures, febrile convulsions and tonic-clonic seizures. The gene that encodes TBC1D24 contains 28,353 bases and maps to human chromosome 16p13.3.

## REFERENCES

1. Zara, F., et al. 2000. Mapping of a locus for a familial autosomal recessive idiopathic myoclonic epilepsy of infancy to chromosome 16p13. *Am. J. Hum. Genet.* 66: 1552-1557.
2. de Curtis, I. 2008. Functions of Rac GTPases during neuronal development. *Dev. Neurosci.* 30: 47-58.
3. Ishibashi, K., et al. 2009. Identification and characterization of a novel Tre-2/Bub2/Cdc16 (TBC) protein that possesses Rab3A-GAP activity. *Genes Cells* 14: 41-52.
4. Falace, A., et al. 2010. TBC1D24, an ARF6-interacting protein, is mutated in familial infantile myoclonic epilepsy. *Am. J. Hum. Genet.* 87: 365-370.

## CHROMOSOMAL LOCATION

Genetic locus: TBC1D24 (human) mapping to 16p13.3; Tbc1d24 (mouse) mapping to 17 A3.3.

## SOURCE

TBC1D24 (G-6) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 437-559 at the C-terminus of TBC1D24 of human origin.

## PRODUCT

Each vial contains 200 µg IgG<sub>1</sub> kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

TBC1D24 (G-6) is available conjugated to agarose (sc-390237 AC), 500 µg/0.25 ml agarose in 1 ml, for IP; to HRP (sc-390237 HRP), 200 µg/ml, for WB, IHC(P) and ELISA; to either phycoerythrin (sc-390237 PE), fluorescein (sc-390237 FITC), Alexa Fluor® 488 (sc-390237 AF488), Alexa Fluor® 546 (sc-390237 AF546), Alexa Fluor® 594 (sc-390237 AF594) or Alexa Fluor® 647 (sc-390237 AF647), 200 µg/ml, for WB (RGB), IF, IHC(P) and FCM; and to either Alexa Fluor® 680 (sc-390237 AF680) or Alexa Fluor® 790 (sc-390237 AF790), 200 µg/ml, for Near-Infrared (NIR) WB, IF and FCM.

Blocking peptide available for competition studies, sc-390237 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

## APPLICATIONS

TBC1D24 (G-6) is recommended for detection of TBC1D24 of mouse, rat and human origin by Western Blotting (starting dilution 1:100, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

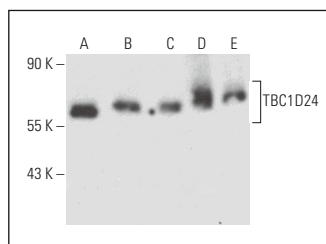
TBC1D24 (G-6) is also recommended for detection of TBC1D24 in additional species, including canine.

Suitable for use as control antibody for TBC1D24 siRNA (h): sc-93059, TBC1D24 siRNA (m): sc-154103, TBC1D24 shRNA Plasmid (h): sc-93059-SH, TBC1D24 shRNA Plasmid (m): sc-154103-SH, TBC1D24 shRNA (h) Lentiviral Particles: sc-93059-V and TBC1D24 shRNA (m) Lentiviral Particles: sc-154103-V.

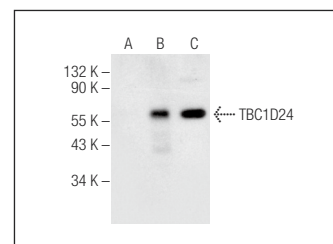
Molecular Weight of TBC1D24: 63 kDa.

Positive Controls: TBC1D24 (h): 293T Lysate: sc-111978, C6 whole cell lysate: sc-364373 or Neuro-2A whole cell lysate: sc-364185.

## DATA



TBC1D24 (G-6): sc-390237. Western blot analysis of TBC1D24 expression in C6 (A), BC<sub>3</sub>H1 (B) and Neuro-2A (C) whole cell lysates and mouse brain (D) and rat brain (E) tissue extracts.



TBC1D24 (G-6): sc-390237. Western blot analysis of TBC1D24 expression in non-transfected 293T: sc-117752 (A), human TBC1D24 transfected 293T: sc-111978 (B) and U-205 (C) whole cell lysates.

## SELECT PRODUCT CITATIONS

1. Campeau, P.M., et al. 2014. The genetic basis of DOORS syndrome: an exome-sequencing study. *Lancet Neurol.* 13: 44-58.
2. Zhang, L., et al. 2014. A dominant mutation in the stereocilia-expressing gene TBC1D24 is a probable cause for nonsyndromic hearing impairment. *Hum. Mutat.* 35: 814-818.
3. Tona, R., et al. 2020. Mouse models of human pathogenic variants of TBC1D24 associated with non-syndromic deafness DFNB86 and DFNA65 and syndromes involving deafness. *Genes* 11: 1122.

## STORAGE

Store at 4° C, \*\*DO NOT FREEZE\*\*. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

## RESEARCH USE

For research use only, not for use in diagnostic procedures.

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