SANTA CRUZ BIOTECHNOLOGY, INC.

RIA1 (A-7): sc-514617

BACKGROUND

RIA1 (ribosome assembly 1), also known as EFTUD1 (elongation factor Tu GTP-binding domain-containing protein 1) or FAM42A, is a 1,120 amino acid protein that exists as three alternatively spliced isoforms and belongs to the GTP-binding elongation factor family. The gene encoding RIA1 maps to human chromosome 15, which is comprised of approximately 106 million base pairs, making up about 3% of the human genome. Angelman and Prader-Willi syndromes are associated with loss of function or deletion of genes in the 15q11-q13 region. In the case of Angelman syndrome, this loss is due to inactivity of the maternal 15q11-q13 encoded UBE3A gene in the brain by either chromosomal deletion or mutation. In cases of Prader-Willi syndrome, there is a partial or complete deletion of this region from the paternal copy of chromosome 15. Tay-Sachs disease is a lethal disorder associated with mutations of the HEXA gene, which is encoded by chromosome 15. Marfan syndrome is associated with chromosome 15 through the FBN1 gene.

REFERENCES

1. Hurowitz, G.I., et al. 1993. Neuropsychiatric aspects of adult-onset Tay-Sachs disease: two case reports with several new findings. J. Neuropsychiatry Clin. Neurosci. 5: 30-36.
2. Zody, M.C., et al. 2006. Analysis of the DNA sequence and duplication history of human chromosome 15. Nature 440: 671-675.
3. Midla, G.S. 2008. Diagnosis and management of patients with Marfan syndrome. JAAPA 21: 21-25.
4. Dan, B. 2009. Angelman syndrome: current understanding and research prospects. Epilepsia 50: 2331-2339.
5. Ferrer-Bolufer, I., et al. 2009. Tyrosinemia type 1 and Angelman syndrome due to paternal uniparental isodisomy 15. J. Inherit. Metab. Dis. 32: S349-S353.
6. Nicolas, E., et al. 2010. CAMOS, a nonprogressive, autosomal recessive, congenital cerebellar ataxia, is caused by a mutant zinc-finger protein, ZNF592. Eur. J. Hum. Genet. 18: 1107-1113.
7. Wawrzik, M., et al. 2010. The C15orf2 gene in the Prader-Willi syndrome region is subject to genomic imprinting and positive selection. Neurogenetics 11: 153-161.

CHROMOSOMAL LOCATION

Genetic locus: EFL1 (human) mapping to 15q25.2; Efl1 (mouse) mapping to 7 D3.

SOURCE

RIA1 (A-7) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 1051-1073 near the C-terminus of RIA1 of human origin.

STORAGE

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

PRODUCT

Each vial contains 200 µg IgG_1 kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-514617 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

APPLICATIONS

RIA1 (A-7) is recommended for detection of RIA1 of human origin and Eftud1 of mouse and rat 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).

Suitable for use as control antibody for RIA1 siRNA (h): sc-90234, Eftud1 siRNA (m): sc-143316, RIA1 shRNA Plasmid (h): sc-90234-SH, Eftud1 shRNA Plasmid (m): sc-143316-SH, RIA1 shRNA (h) Lentiviral Particles: sc-90234-V and Eftud1 shRNA (m) Lentiviral Particles: sc-143316-V.

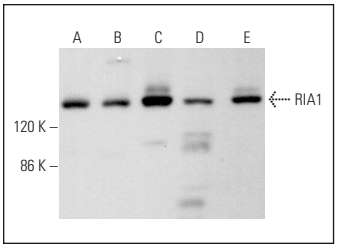
Molecular Weight of RIA1 isoforms: 125/120/96 kDa.

Positive Controls: RIA1 (h): 293T Lysate: sc-172214, HeLa whole cell lysate: sc-2200 or NIH/3T3 whole cell lysate: sc-2210.

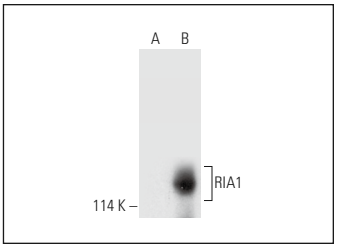
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

DATA



RIA1 (A-7): sc-514617. Western blot analysis of RIA1 expression in HeLa (**A**), A549 (**B**), NIH/3T3 (**C**), SP2/0 (**D**) and RPE-J (**E**) whole cell lysates.



RIA1 (A-7): sc-514617. Western blot analysis of RIA1 expression in non-transfected: sc-117752 (**A**) and human RIA1 transfected: sc-172214 (**B**) 293T whole cell lysates.

RESEARCH USE

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