

# RIA1 (C-15): sc-240829

## 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

- Hurowitz, G.I., Silver, J.M., Brin, M.F., Williams, D.T. and Johnson, W.G. 1993. Neuropsychiatric aspects of adult-onset Tay-Sachs disease: two case reports with several new findings. *J. Neuropsychiatry Clin. Neurosci.* 5: 30-36.
- Zody, M.C., Garber, M., Sharpe, T., Young, S.K., Rowen, L., O'Neill, K., Whittaker, C.A., Kamal, M., Chang, J.L., Cuomo, C.A., Dewar, K., FitzGerald, M.G., Kodira, C.D., Madan, A., Qin, S., Yang, X., Abbasi, N., Abouelleil, A., Arachchi, H.M., et al. 2006. Analysis of the DNA sequence and duplication history of human chromosome 15. *Nature* 440: 671-675.
- Midla, G.S. 2008. Diagnosis and management of patients with Marfan syndrome. *JAAPA* 21: 21-25.
- Dan, B. 2009. Angelman syndrome: current understanding and research prospects. *Epilepsia* 50: 2331-2339.
- Ferrer-Bolufer, I., Dalmau, J., Quiroga, R., Oltra, S., Orellana, C., Monfort, S., Roselló, M., De La Osa, A. and Martinez, F. 2009. Tyrosinemia type 1 and Angelman syndrome due to paternal uniparental isodisomy 15. *J. Inherit. Metab. Dis.* 32: S349-S353.
- Nicolas, E., Poitelon, Y., Chouery, E., Salem, N., Levy, N., Megarbane, A. and Delague, V. 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.
- Wawrzik, M., Unmehopa, U.A., Swaab, D.F., van de Nes, J., Buiting, K. and Horsthemke, B. 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: EFTUD1 (human) mapping to 15q25.2; Eftud1 (mouse) mapping to 7 D3.

## SOURCE

RIA1 (C-15) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the C-terminus of RIA1 of human origin.

## PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-240829 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

RIA1 (C-15) is recommended for detection of Eftud1 of mouse and rat origin, and RIA1 of human origin of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), 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).

RIA1 (C-15) is also recommended for detection of RIA1 in additional species, including equine, canine, bovine, porcine and avian.

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.

## RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

## 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.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) or our catalog for detailed protocols and support products.