

RIA1 (h): 293T Lysate: sc-172214

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 3 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

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- Midla, G.S. 2008. Diagnosis and management of patients with Marfan syndrome. *JAAPO* 21: 21-25.
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- Ferrer-Boluer, I., et al. 2009. Tyrosinemia type 1 and Angelman syndrome due to paternal uniparental isodisomy 15. *J. Inher. Metab. Dis.* 32: S349-S353.
- 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.
- 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.

PRODUCT

RIA1 (h): 293T Lysate represents a lysate of human RIA1 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

APPLICATIONS

RIA1 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive RIA1 antibodies. Recommended use: 10-20 µl per lane.

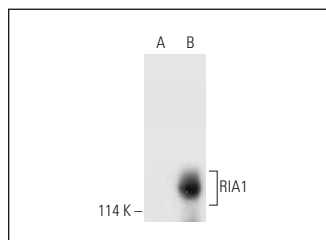
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

RIA1 (A-7): sc-514617 is recommended as a positive control antibody for Western Blot analysis of enhanced human RIA1 expression in RIA1 transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

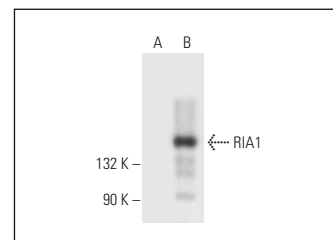
RECOMMENDED SUPPORT REAGENTS

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.

DATA



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.



RIA1 (C-5): sc-398435. 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.

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.