

ATXN7L2 (h): 293T Lysate: sc-115673

BACKGROUND

SCA7 is an autosomal dominant neurodegenerative disorder characterized by ataxia and selective neuronal cell loss caused by the expansion of a translated CAG repeat encoding a polyglutamine tract in ataxin-7, which is the SCA7 gene product. Ataxin-7 is a nuclear protein that is expressed within neurons both affected and unaffected in SCA7 pathology with subcellular localization being variable depending upon the neuronal subtype. Polyglutamine expanded in ataxin-7 may carry out its pathogenic effects in the nucleus by altering the matrix-associated nuclear structure and/or by disrupting nucleolar function. ATXN7L2 (Ataxin-7-like protein 2) is a 722 amino acid protein that contains a SCA7 domain, which is highly conserved through all members of the ATXN7 gene family. The gene encoding ATXN7L2 maps to human chromosome 1p13.3, the largest human chromosome spanning about 260 million base pairs and making up 8% of the human genome.

REFERENCES

1. Holmberg, M., et al. 1998. Spinocerebellar ataxia type 7 (SCA7): a neurodegenerative disorder with neuronal intranuclear inclusions. *Hum. Mol. Genet.* 7: 913-918.
2. Chen, S., et al. 2004. Interference of Crx-dependent transcription by ataxin-7 involves interaction between the glutamine regions and requires the ataxin-7 carboxy-terminal region for nuclear localization. *Hum. Mol. Genet.* 13: 53-67.
3. Gregory, S.G., et al. 2006. The DNA sequence and biological annotation of human chromosome 1. *Nature* 441: 315-321.
4. Garden, G.A. and La Spada, A.R. 2008. Molecular pathogenesis and cellular pathology of spinocerebellar ataxia type 7 neurodegeneration. *Cerebellum* 7: 138-149.
5. Zhao, Y., et al. 2008. A TERC/STAGA module mediates histone H2A and H2B deubiquitination, coactivates nuclear receptors, and counteracts heterochromatin silencing. *Mol. Cell* 29: 92-101.
6. Lin, Y., et al. 2008. Trinucleotide expansions in the SCA7 gene in a large family with spinocerebellar ataxia and craniocervical dystonia. *Neurosci. Lett.* 434: 230-233.
7. Hugosson, T., et al. 2009. Macular dysfunction and morphology in spinocerebellar ataxia type 7 (SCA 7). *Ophthalmic Genet.* 30: 1-6.
8. Chou, A.H., et al. 2010. Polyglutamine-expanded ataxin-7 causes cerebellar dysfunction by inducing transcriptional dysregulation. *Neurochem. Int.* 56: 329-339.

CHROMOSOMAL LOCATION

Genetic locus: ATXN7L2 (human) mapping to 1p13.3.

PRODUCT

ATXN7L2 (h): 293T Lysate represents a lysate of human ATXN7L2 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

RESEARCH USE

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

APPLICATIONS

ATXN7L2 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive ATXN7L2 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.

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.

PROTOCOLS

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