

# C1orf70 siRNA (h): sc-88115

## BACKGROUND

C1orf70, also known as TMEM240 (Transmembrane protein 240) or SCA21 (Spinocerebellar ataxia 21), is a 173 amino acid transmembrane protein that belongs to the TMEM240 family. Mutations in the gene encoding C1orf70 causes Spinocerebellar ataxia 21 (SCA21). SCA21 is disease characterized by incoordination of gait, speech and eye movements. The gene encoding C1orf70 maps to human chromosome 1, which is the largest human chromosome spanning about 260 million base pairs and makes up 8% of the human genome. There are about 3,000 genes on chromosome 1, and considering the great number of genes there are also a large number of diseases associated with chromosome 1. Notably, the rare aging disease Hutchinson-Gilford progeria is associated with the LMNA gene which encodes lamin A. When defective, the LMNA gene product can build up in the nucleus and cause characteristic nuclear blebs. The mechanism of rapidly enhanced aging is unclear and is a topic of continuing exploration. The MUTYH gene is located on chromosome 1 and is partially responsible for familial adenomatous polyposis. Stickler syndrome, Parkinsons, Gaucher disease and Usher syndrome are also associated with chromosome 1. A breakpoint has been identified in 1q which disrupts the DISC1 gene and is linked to schizophrenia. Aberrations in chromosome 1 are found in a variety of cancers including head and neck cancer, malignant melanoma and multiple myeloma.

## REFERENCES

1. Watson, M.L., et al. 1990. Genomic organization of the selectin family of leukocyte adhesion molecules on human and mouse chromosome 1. *J. Exp. Med.* 172: 263-272.
2. Blackwood, D.H., et al. 2001. Schizophrenia and affective disorders— cosegregation with a translocation at chromosome 1q42 that directly disrupts brain-expressed genes: clinical and P300 findings in a family. *Am. J. Hum. Genet.* 69: 428-433.
3. Gregory, S.G., et al. 2006. The DNA sequence and biological annotation of human chromosome 1. *Nature* 441: 315-321.
4. Hennah, W., et al. 2006. Genes and schizophrenia: beyond schizophrenia: the role of DISC1 in major mental illness. *Schizophr. Bull.* 32: 409-416.
5. Lans, H. and Hoeijmakers, J.H. 2006. Cell biology: aging nucleus gets out of shape. *Nature* 440: 32-34.
6. Marzin, Y., et al. 2006. Chromosome 1 abnormalities in multiple myeloma. *Anticancer Res.* 26: 953-959.
7. McClintock, D., et al. 2006. Hutchinson-Gilford progeria mutant Lamin A primarily targets human vascular cells as detected by an anti-Lamin A G608G antibody. *Proc. Natl. Acad. Sci. USA* 103: 2154-2159.
8. Scaffidi, P. and Misteli, T. 2006. Lamin A-dependent nuclear defects in human aging. *Science* 312: 1059-1063.
9. Delplanque, J., et al. 2014. TMEM240 mutations cause spinocerebellar ataxia 21 with mental retardation and severe cognitive impairment. *Brain* 137: 2657-2663.

## CHROMOSOMAL LOCATION

Genetic locus: TMEM240 (human) mapping to 1p36.33.

## PRODUCT

C1orf70 siRNA (h) is a pool of 2 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10  $\mu$ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see C1orf70 shRNA Plasmid (h): sc-88115-SH and C1orf70 shRNA (h) Lentiviral Particles: sc-88115-V as alternate gene silencing products.

For independent verification of C1orf70 (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-88115A and sc-88115B.

## STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNAses and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330  $\mu$ l of the RNase-free water provided. Resuspension of the siRNA duplex in 330  $\mu$ l of RNase-free water makes a 10  $\mu$ M solution in a 10  $\mu$ M Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

## APPLICATIONS

C1orf70 siRNA (h) is recommended for the inhibition of C1orf70 expression in human cells.

## SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10  $\mu$ M in 66  $\mu$ l. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

## RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor C1orf70 gene expression knockdown using RT-PCR Primer: C1orf70 (h)-PR: sc-88115-PR (20  $\mu$ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

## 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) for detailed protocols and support products.