C4orf48 (N-12): sc-245792



The Power to Question

BACKGROUND

Representing approximately 6% of the human genome, chromosome 4 contains nearly 900 genes. Notably, the Huntingtin gene, which is found to encode an expanded glutamine tract in cases of Huntington's disease, is on chromosome 4. FGFR-3 is also encoded on chromosome 4 and has been associated with thanatophoric dwarfism, achondroplasia, Muenke syndrome and bladder cancer. Chromosome 4 is also tied to Ellis-van Creveld syndrome, methylmalonic acidemia and polycystic kidney disease. Chromosome 4 reportedly contains the largest gene deserts (regions of the genome with no protein encoding genes) and has one of the two lowest recombination frequencies of the human chromosomes. The C4orf48 gene product has been provisionally designated C4orf48 pending further characterization.

REFERENCES

- 1. Hillier, L.W., Graves, T.A., Fulton, R.S., Fulton, L.A., Pepin, K.H., Minx, P., Wagner-McPherson, C., Layman, D., Wylie, K., Sekhon, M., Becker, M.C., Fewell, G.A., et al. 2005. Generation and annotation of the DNA sequences of human chromosomes 2 and 4. Nature 434: 724-731.
- 2. Cowan, C.M. and Raymond, L.A. 2006. Selective neuronal degeneration in Huntington's disease. Curr. Top. Dev. Biol. 75: 25-71.
- Chandler, R.J., Sloan, J., Fu, H., Tsai, M., Stabler, S., Allen, R., Kaestner, K.H., Kazazian, H.H. and Venditti, C.P. 2007. Metabolic phenotype of methylmalonic acidemia in mice and humans: the role of skeletal muscle. BMC Med. Genet. 8: 64
- Cunningham, M.L., Seto, M.L., Ratisoontorn, C., Heike, C.L. and Hing, A.V. 2007. Syndromic craniosynostosis: from history to hydrogen bonds. Orthod. Craniofac. Res. 10: 67-81.
- 5. de Frutos, C.A., Vega, S., Manzanares, M., Flores, J.M., Huertas, H., Martínez-Frías, M.L. and Nieto, M.A. 2007. Snail1 Is a transcriptional effector of FGFR-3 signaling during chondrogenesis and achondroplasias. Dev. Cell 13: 872-883.
- Ruiz-Perez, V.L., Blair, H.J., Rodriguez-Andres, M.E., Blanco, M.J., Wilson, A., Liu, Y.N., Miles, C., Peters, H. and Goodship, J.A. 2007. EVC is a positive mediator of lhh-regulated bone growth that localises at the base of chondrocyte cilia. Development 134: 2903-2912.
- Stack, E.C., Dedeoglu, A., Smith, K.M., Cormier, K., Kubilus, J.K., Bogdanov, M., Matson, W.R., Yang, L., Jenkins, B.G., Luthi-Carter, R., Kowall, N.W., Hersch, S.M., Beal, M.F. and Ferrante, R.J. 2007. Neuroprotective effects of synaptic modulation in Huntington's disease R6/2 mice. J. Neurosci. 27: 12908-12915.
- Versteegh, F.G., Buma, S.A., Costin, G., de Jong, W.C. and Hennekam, R.C. 2007. EvC Working Party. Growth hormone analysis and treatment in Ellisvan Creveld syndrome. Am. J. Med. Genet. A 143: 2113-2121.
- Doherty, E.S., Lacbawan, F., Hadley, D.W., Brewer, C., Zalewski, C., Kim, H.J., Solomon, B., Rosenbaum, K., Domingo, D.L., Hart, T.C., Brooks, B.P., Immken, L., Lowry, R.B., Kimonis, V., Shanske, A.L., Jehee, F.S., et al. 2007. Muenke syndrome (FGFR-3-related craniosynostosis): Expansion of the phenotype and review of the literature. Am. J. Med. Genet. A 143: 3204-3215.

CHROMOSOMAL LOCATION

Genetic locus: C4orf48 (human) mapping to 4p16.3.

SOURCE

C4orf48 (N-12) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the N-terminus of C4orf48 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-245792 P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

C4orf48 (N-12) is recommended for detection of C4orf48 of 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).

Suitable for use as control antibody for C4orf48 siRNA (h): sc-89147, C4orf48 shRNA Plasmid (h): sc-89147-SH and C4orf48 shRNA (h) Lentiviral Particles: sc-89147-V.

Molecular Weight of C4orf48: 14 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) 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 or our catalog for detailed protocols and support products.

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