FAM47E (C-20): sc-246729



The Power to Question

BACKGROUND

FAM47E is a 351 amino acid protein that is encoded by a gene that maps to human chromosome 4, that represents approximately 6% of the human genome and 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.

REFERENCES

- 1. Hillier, L.W., 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., et al. 2007. Metabolic phenotype of methylmalonic acidemia in mice and humans: the role of skeletal muscle. BMC Med. Genet. 8: 64.
- 4. Cunningham, M.L., et al. 2007. Syndromic craniosynostosis: from history to hydrogen bonds. Orthod. Craniofac. Res. 10: 67-81.
- de Frutos, C.A., et al. 2007. Snail1 is a transcriptional effector of FGFR-3 Signaling during chondrogenesis and achondroplasias. Dev. Cell 13: 872-883.
- Doherty, E.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.
- Ruiz-Perez, V.L., et al. 2007. EVC is a positive mediator of lhh-regulated bone growth that localises at the base of chondrocyte cilia. Development 134: 2903-2912.
- 8. Versteegh, F.G., et al. 2007. Growth hormone analysis and treatment in Ellis-van Creveld syndrome. Am. J. Med. Genet. A 143: 2113-2121.

CHROMOSOMAL LOCATION

Genetic locus: FAM47E (human) mapping to 4q21.1.

SOURCE

FAM47E (C-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of FAM47E of human origin.

PRODUCT

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

Blocking peptide available for competition studies, sc-246729 P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

FAM47E (C-20) is recommended for detection of FAM47E 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); non cross-reactive with FAM47A, FAM47B or FAM47C.

Molecular Weight of FAM47E isoforms: 41/35 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 or our catalog for detailed protocols and support products.

Santa Cruz Biotechnology, Inc. 1.800.457.3801 831.457.3801 **Europe** +00800 4573 8000 49 6221 4503 0 **www.scbt.com**