

# CHCHD2 (K-19): sc-246277

## BACKGROUND

CHCHD2 (coiled-coil-helix-coiled-coil-helix domain containing 2), also known as C7orf17, is a 151 amino acid mitochondrial protein containing one CHCH domain and is encoded by a gene located on human chromosome 7. Chromosome 7 is approximately 158 million bases in length, encodes over 1000 genes and makes up about 5% of the human genome. Chromosome 7 has been linked to Osteogenesis imperfecta, Pendred syndrome, Lissencephaly, Citrullinemia and Shwachman-Diamond syndrome. The deletion of a portion of the q arm of chromosome 7 is associated with Williams-Beuren syndrome, a condition characterized by mild mental retardation, an unusual comfourt and friendliness with strangers and an elfin appearance. Deletions of portions of the q arm of chromosome 7 are also seen in a number of myeloid disorders including cases of acute myelogenous leukemia and myelodysplasia.

## REFERENCES

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3. Hillier, L.W., et al. 2003. The DNA sequence of human chromosome 7. *Nature* 424: 157-164.
4. Eckert, M.A., et al. 2006. The neurobiology of Williams syndrome: cascading influences of visual system impairment? *Cell. Mol. Life Sci.* 63: 1867-1875.
5. Osborne, L.R., et al. 2006. Williams-Beuren syndrome diagnosis using fluorescence *in situ* hybridization. *Methods Mol. Med.* 126:113-128.
6. Reiner, O., et al. 2006. Lissencephaly 1 linking to multiple diseases: mental retardation, neurodegeneration, schizophrenia, male sterility, and more. *Neuromolecular Med.* 8: 547-565.
7. Shimamura, A. 2006. Shwachman-Diamond syndrome. *Semin. Hematol.* 43: 178-188.
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## CHROMOSOMAL LOCATION

Genetic locus: Chchd2/2410018M08Rik (mouse) mapping to 5 G1.3.

## SOURCE

CHCHD2 (K-19) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of CHCHD2 of mouse origin.

## STORAGE

Store at 4° C, **\*\*DO NOT FREEZE\*\***. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

## PRODUCT

Each vial contains 200  $\mu$ g IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-246277 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

CHCHD2 (K-19) is recommended for detection of CHCHD2 and Scand3 of mouse and rat 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).

Molecular Weight of CHCHD2: 16 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.

## 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) or our catalog for detailed protocols and support products.