# SANTA CRUZ BIOTECHNOLOGY, INC.

# VPS13A (C-13): sc-109138



# BACKGROUND

Vacuolar sorting proteins (VPSs) are required for proper trafficking of endocytic and biosynthetic proteins to the vacuole and play an important role in the budding process of cells. VPS13A (vacuolar protein sorting 13 homolog A), also known as CHOC or CHOREIN, is a 3,174 amino acid protein that belongs to the VPS family and contains 10 TPR repeats. Expressed in a variety of tissues, including brain, kidney, heart and skeletal muscle, VPS13A is thought to play a role in the regulation of protein cycling from the Golgi network to endosomes, lysosomes and the plasma membrane. Defects in the gene encoding VPS13A are the cause of chorea-acanthocytosis (CHAC), an autosomal recessive disorder characterized by epilepsy, peripheral neuropathy, myopathy and oral selfmutilation. Multiple isoforms of VPS13A exist due to alternative splicing events.

### REFERENCES

- 1. Rubio, J.P., et al. 1997. Chorea-acanthocytosis: genetic linkage to chromosome 9q21. Am. J. Hum. Genet. 61: 899-908.
- Rampoldi, L., et al. 2001. A conserved sorting-associated protein is mutant in chorea-acanthocytosis. Nat. Genet. 28: 119-120.
- Ueno, S., et al. 2001. The gene encoding a newly discovered protein, CHOREIN, is mutated in chorea-acanthocytosis. Nat. Genet. 28: 121-122.
- Bohlega, S., et al. 2003. Chorea-acanthocytosis: clinical and genetic findings in three families from the Arabian peninsula. Mov. Disord. 18: 403-407.
- Velayos-Baeza, A., et al. 2004. Analysis of the human VPS13 gene family. Genomics 84: 536-549.
- Dobson-Stone, C., et al. 2005. Identification of a VPS13A founder mutation in French Canadian families with chorea-acanthocytosis. Neurogenetics 6: 151-158.
- 7. Ruiz-Sandoval, J.L., et al. 2007. Choreoacanthocytosis in a Mexican family. Arch. Neurol. 64: 1661-1664.
- 8. Ichiba, M., et al. 2007. Clinical and molecular genetic assessment of a chorea-acanthocytosis pedigree. J. Neurol. Sci. 263: 124-132.

# CHROMOSOMAL LOCATION

Genetic locus: VPS13A (human) mapping to 9q21.2; Vps13a (mouse) mapping to 19 B.

#### SOURCE

VPS13A (C-13) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of VPS13A of human origin.

### 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-109138 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

### APPLICATIONS

VPS13A (C-13) is recommended for detection of VPS13A of mouse, rat and 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).

PS13A (C-13) is also recommended for detection of VPS13A in additional species, including canine and bovine.

Suitable for use as control antibody for VPS13A siRNA (h): sc-92870, VPS13A siRNA (m): sc-155217, VPS13A shRNA Plasmid (h): sc-92870-SH, VPS13A shRNA Plasmid (m): sc-155217-SH, VPS13A shRNA (h) Lentiviral Particles: sc-92870-V and VPS13A shRNA (m) Lentiviral Particles: sc-155217-V.

Molecular Weight of VPS13A: 360 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) Immunofluo-rescence: 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.