

# SLC48A1 (B-16): sc-101957

## BACKGROUND

SLC48A1 is a 239 amino acid protein that is encoded by a gene located on chromosome 12. Encoding over 1,100 genes within 132 million bases, chromosome 12 makes up about 4.5% of the human genome. A number of skeletal deformities are linked to chromosome 12 including hypochondrogenesis, achondrogenesis and Kniest dysplasia. Noonan syndrome, which includes heart and facial developmental defects among the primary symptoms, is caused by a mutant form of PTPN11 gene product, SH-PTP2. Chromosome 12 is also home to a homeobox gene cluster which encodes crucial transcription factors for morphogenesis, and the natural killer complex gene cluster encoding C-type lectin proteins which mediate the NK cell response to MHC I interaction. Trisomy 12p leads to facial development defects, seizure disorders and a host of other symptoms varying in severity depending on the extent of mosaicism and is most severe in cases of complete trisomy.

## REFERENCES

- Allen, T.L., et al. 1996. Cytogenetic and molecular analysis in trisomy 12p. *Am. J. Med. Genet.* 63: 250-256.
- Yang, W. and Cole, W.G. 1998. Low basal transcripts of the COL2A1 collagen gene from lymphoblasts show alternative splicing of exon 12 in the Kniest form of spondyloepiphyseal dysplasia. *Hum. Mutat.* 1: 1-2.
- Trowsdale, J., et al. 2001. The genomic context of natural killer receptor extended gene families. *Immunol. Rev.* 181: 20-38.
- Zumkeller, W., et al. 2004. Genotype/phenotype analysis in a patient with pure and complete trisomy 12p. *Am. J. Med. Genet. A* 129: 261-264.
- Kelley, J., et al. 2005. Comparative genomics of natural killer cell receptor gene clusters. *PLoS Genet.* 1: e27.
- Nishimura, G., et al. 2005. The phenotypic spectrum of COL2A1 mutations. *Hum. Mutat.* 26: 36-43.
- Segel, R., et al. 2006. The natural history of trisomy 12p. *Am. J. Med. Genet. A* 140: 695-703.

## CHROMOSOMAL LOCATION

Genetic locus: SLC48A1 (human) mapping to 12q13.11.

## SOURCE

SLC48A1 (B-16) is a purified rabbit polyclonal antibody raised against SLC48A1 of human origin.

## PRODUCT

Each vial contains 50 µg IgG in 500 µl PBS with < 0.1% sodium azide, 0.1% gelatin and < 0.02% sucrose.

## 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.

## APPLICATIONS

SLC48A1 (B-16) is recommended for detection of SLC48A1 of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500), immunohistochemistry (including paraffin-embedded sections) (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 SLC48A1 siRNA (h): sc-95722, SLC48A1 shRNA Plasmid (h): sc-95722-SH and SLC48A1 shRNA (h) Lentiviral Particles: sc-95722-V.

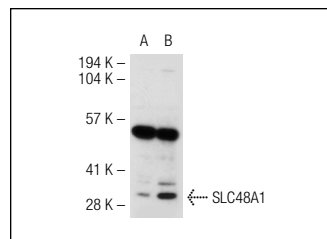
Molecular Weight of SLC48A1: 30 kDa.

Positive Controls: SLC48A1 (h): 293T Lysate: sc-117109, Jurkat whole cell lysate: sc-2204 or Hep G2 cell lysate: sc-2227.

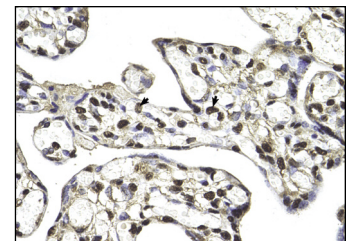
## RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941. 4) Immunohistochemistry: use ImmunoCruz™: sc-2051 or ABC: sc-2018 rabbit IgG Staining Systems.

## DATA



SLC48A1 (B-16): sc-101957. Western blot analysis of SLC48A1 expression in non-transfected: sc-117752 (A) and human SLC48A1 transfected: sc-117109 (B). 293T whole cell lysates.



SLC48A1 (B-16): sc-101957. Immunoperoxidase staining of formalin fixed, paraffin-embedded human placenta tissue showing nuclear and cytoplasmic staining.

## SELECT PRODUCT CITATIONS

- Campbell, M.R., et al. 2013. Novel hematopoietic target genes in the NRF2-mediated transcriptional pathway. *Oxid. Med. Cell. Longev.* 2013: 120305.
- Hooda, J., et al. 2013. Enhanced heme function and mitochondrial respiration promote the progression of lung cancer cells. *PLoS ONE* 8: e63402.