

OPA3 (P-20): sc-82006

BACKGROUND

OPA3 (optic atrophy 3), also known as MGA3, is a 179 amino acid mitochondrial protein that is highly expressed in kidney and skeletal muscle and is thought to play a role in a variety of mitochondrial processes. Defects in the gene encoding OPA3 are the cause of both 3-methylglutaconic aciduria type 3 (MGA3) and optic atrophy type 3 (OPA3). MGA3, also known as optic atrophy plus syndrome or Costeff optic atrophy syndrome, is an autosomal recessive disorder that is characterized by early-onset bilateral optic atrophy, spasticity, extrapyramidal dysfunction and cognitive deficit. OPA3, a neurological disorder, is also characterized by visual impairment, including severe loss of visual acuity, temporal optic disk pallor, color vision deficits and centrocecal scotoma of variable density. Multiple isoforms of OPA3 exist due to alternative splicing events.

REFERENCES

1. Nystuen, A., Costeff, H., Elpeleg, O.N., Apter, N., Bonne-Tamir, B., Mohrenweiser, H., Haider, N., Stone, E.M. and Sheffield, V.C. 1997. Iraqi-Jewish kindreds with optic atrophy plus (3-methylglutaconic aciduria type 3) demonstrate linkage disequilibrium with the CTG repeat in the 3' untranslated region of the myotonic dystrophy protein kinase gene. *Hum. Mol. Genet.* 6: 563-569.
2. Anikster, Y., Kleta, R., Shaag, A., Gahl, W.A. and Elpeleg, O. 2001. Type III 3-methylglutaconic aciduria (optic atrophy plus syndrome, or Costeff optic atrophy syndrome): identification of the OPA3 gene and its founder mutation in Iraqi Jews. *Am. J. Hum. Genet.* 69: 1218-1224.
3. Kleta, R., Skovby, F., Christensen, E., Rosenberg, T., Gahl, W.A. and Anikster, Y. 2002. 3-Methylglutaconic aciduria type III in a non-Iraqi-Jewish kindred: clinical and molecular findings. *Mol. Genet. Metab.* 76: 201-206.
4. Reynier, P., Amati-Bonneau, P., Verry, C., Olichon, A., Simard, G., Guichet, A., Bonnemains, C., Malecaze, F., Malinge, M.C., Pelletier, J.B., Calvas, P., Dollfus, H., Belenguer, P., Malthiery, Y., Lenaers, G. and Bonneau, D. 2004. OPA3 gene mutations responsible for autosomal dominant optic atrophy and cataract. *J. Med. Genet.* 41: e110.
5. Fink, N. and Mouallem, M. 2006. Costeff syndrome: a syndrome that was described in Israel and the responsible gene discovered by an Israeli doctor. *Harefuah.* 145: 402-403, 472.
6. Online Mendelian Inheritance in Man, OMIM™. 2007. Johns Hopkins University, Baltimore, MD. MIM Number: 606580. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
7. Chevrollier, A., Guillet, V., Loiseau, D., Gueguen, N., de Crescenzo, M.A., Verry, C., Ferre, M., Dollfus, H., Odent, S., Milea, D., Goizet, C., Amati-Bonneau, P., Procaccio, V., Bonneau, D. and Reynier, P. 2008. Hereditary optic neuropathies share a common mitochondrial coupling defect. *Ann. Neurol.* 63: 794-798.

CHROMOSOMAL LOCATION

Genetic locus: OPA3 (human) mapping to 19q13.32; Opa3 (mouse) mapping to 7 A3.

SOURCE

OPA3 (P-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of OPA3 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-82006 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

OPA3 (P-20) is recommended for detection of OPA3 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).

OPA3 (P-20) is also recommended for detection of OPA3 in additional species, including canine.

Suitable for use as control antibody for OPA3 siRNA (h): sc-75994, OPA3 siRNA (m): sc-75995, OPA3 shRNA Plasmid (h): sc-75994-SH, OPA3 shRNA Plasmid (m): sc-75995-SH, OPA3 shRNA (h) Lentiviral Particles: sc-75994-V and OPA3 shRNA (m) Lentiviral Particles: sc-75995-V.

Molecular Weight of OPA3: 20 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.