

# Twinkle (D-16): sc-55415

## BACKGROUND

Twinkle, also known as PEO1 (progressive external ophthalmoplegia 1 protein), PEOA3, SANDO or TWINKL, is a mitochondrial protein that functions as a 5'-3' nucleotide-dependent DNA helicase. co-localized with mtDNA (mitochondrial DNA) in mitochondrial nucleoids, Twinkle is important in the metabolism and maintenance of mtDNA, playing a crucial role in the regulation of mtDNA copy numbers. Twinkle is expressed at high levels in testis, pancreas and skeletal muscle and exists as three isoforms due to alternative splicing events. Defects in the gene encoding Twinkle are the cause of two conditions: progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal dominant 3 (PEOA3) and sensory ataxic neuropathy dysarthria and ophthalmoparesis (SANDO). PEOA3 is characterized by ptosis and weak muscles, while SANDO is characterized by ophthalmoparesis, dysarthria and sensory ataxic neuropathies.

## REFERENCES

1. Korhonen, J.A., et al. 2003. TWINKLE Has 5' → 3' DNA helicase activity and is specifically stimulated by mitochondrial single-stranded DNA-binding protein. *J. Biol. Chem.* 278: 48627-48632.
2. Wanrooij, S., et al. 2004. Twinkle and POLG defects enhance age-dependent accumulation of mutations in the control region of mtDNA. *Nucleic Acids Res.* 32: 3053-3064.
3. Tynismaa, H., et al. 2004. Twinkle helicase is essential for mtDNA maintenance and regulates mtDNA copy number. *Hum. Mol. Genet.* 13: 3219-3227.
4. Timmons, J.A., et al. 2006. Expression profiling following local muscle inactivity in humans provides new perspective on diabetes-related genes. *Genomics* 87: 165-172.
5. Wanrooij, S., et al. 2007. Expression of catalytic mutants of the mtDNA helicase Twinkle and polymerase POLG causes distinct replication stalling phenotypes. *Nucleic Acids Res.* 35: 3238-3251.
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7. Baloh, R.H., et al. 2007. Familial parkinsonism and ophthalmoplegia from a mutation in the mitochondrial DNA helicase twinkle. *Arch. Neurol.* 64: 998-1000.
8. Sarzi, E., et al. 2007. Twinkle helicase (PEO1) gene mutation causes mitochondrial DNA depletion. *Ann. Neurol.* 62: 579-587.

## CHROMOSOMAL LOCATION

Genetic locus: C10orf2 (human) mapping to 10q24.31; Peo1 (mouse) mapping to 19 C3.

## SOURCE

Twinkle (D-16) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of Twinkle 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-55415 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

Available as TransCruz reagent for Gel Supershift and ChIP applications, sc-55415 X, 200 µg/0.1 ml.

## APPLICATIONS

Twinkle (D-16) is recommended for detection of Twinkle 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).

Twinkle (D-16) is also recommended for detection of Twinkle in additional species, including equine, canine, bovine, porcine and avian.

Suitable for use as control antibody for Twinkle siRNA (h): sc-63177, Twinkle siRNA (m): sc-63178, Twinkle shRNA Plasmid (h): sc-63177-SH, Twinkle shRNA Plasmid (m): sc-63178-SH, Twinkle shRNA (h) Lentiviral Particles: sc-63177-V and Twinkle shRNA (m) Lentiviral Particles: sc-63178-V.

Twinkle (D-16) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

Molecular Weight of Twinkle: 77 kDa.

Positive Controls: Jurkat whole cell lysate: sc-2204 or Sol8 cell lysate: sc-2249.

## 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 Blotting 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](http://www.scbt.com) or our catalog for detailed protocols and support products.