SANTA CRUZ BIOTECHNOLOGY, INC.

Twinkle (1C5): sc-293368

SANTA CRUZ BIOTECHNOLOGY

The Power to Question

BACKGROUND

Twinkle, also known as PEO1 (progressive external ophthalmoplegia 1 protein), PEOA3, SANDO or TWINL, 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. Tyynismaa, 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.
6. Rivera, H., et al. 2007. Mild ocular myopathy associated with a novel mutation in mitochondrial Twinkle helicase. Neuromuscul. Disord. 17: 677-680.
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.
9. Hakonen, A.H., et al. 2007. Recessive Twinkle mutations in early onset encephalopathy with mtDNA depletion. Brain 130: 3032-3040.

CHROMOSOMAL LOCATION

Genetic locus: C10orf2 (human) mapping to 10q24.31.

SOURCE

Twinkle (1C5) is a mouse monoclonal antibody raised against amino acids 591-684 of Twinkle of human origin.

PRODUCT

Each vial contains 100 µg IgG_{2a} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

Twinkle (1C5) is recommended for detection of Twinkle 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)] and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

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

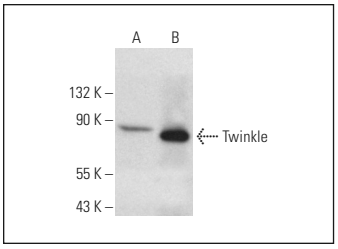
Molecular Weight of Twinkle: 77 kDa.

Positive Controls: human skeletal muscle extract: sc-363776 or Jurkat nuclear extract: sc-2132.

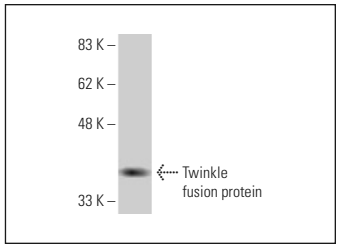
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml).

DATA



Twinkle (1C5): sc-293368. Western blot analysis of Twinkle expression in Jurkat nuclear extract (**A**) and human skeletal muscle tissue extract (**B**).



Twinkle (1C5): sc-293368. Western blot analysis of human recombinant Twinkle fusion protein.

SELECT PRODUCT CITATIONS

1. Mishra, A., et al. 2018. RAD51C/XRCC3 facilitates mitochondrial DNA replication and maintains integrity of the mitochondrial genome. Mol. Cell. Biol. 38: e00489-17.

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.