SANTA CRUZ BIOTECHNOLOGY, INC.

dysferlin (C-19): sc-16635

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The Power to Question

BACKGROUND

Dysferlin is a muscle-specific protein that is essential for normal muscle function and development. Mutations in the human dysferlin gene, DYSF, which maps to chromosome 2p13.2, are associated with limb girdle muscular dystrophy-2B (LGMD-2B) and a related, adult-onset, distal dystrophy known as Miyoshi myopathy (MM). Dysferlin localizes to the muscle fiber membrane, but is absent in MM and LGMD-2B muscle. Dysferlin is detected in 5-6 week embryos, when limbs begin to form regional differentiation. Although it is not essential for initial myogenesis, dysferlin appears to be critical for sustained normal function in mature muscle. It has been suggested that the absence of dysferlin during development gives rise to the disease phenotype in adulthood. Identical mutations in the dysferlin gene can produce more than one myopathy phenotype, indicating that additional genes and/or other factors are also involved in the clinical phenotype. The DYSF gene has no homology to any other known mammalian gene, but the protein product is related to the spermatogenesis factor fer-1 of *Caenorhabditis elegans*. The name 'dysferlin' combines the role of the gene in producing muscular dystrophy with its homology to *C. elegans*.

REFERENCES

1. Bashir, R., et al. 1994. A gene for autosomal recessive limb-girdle muscular dystrophy maps to chromosome 2p. Hum. Mol. Genet. 3: 455-457.
2. Liu, J., et al. 1998. Dysferlin, a novel skeletal muscle gene, is mutated in Miyoshi myopathy and limb girdle muscular dystrophy. Nat. Genet. 20: 31-36.
3. Matsuda, C., et al. 1999. Dysferlin is a surface membrane-associated protein that is absent in Miyoshi myopathy. Neurology 53: 1119-1122.
4. Anderson, L.V., et al. 1999. Dysferlin is a plasma membrane protein and is expressed early in human development. Hum. Mol. Genet. 8: 855-861.
5. Weiler, T., et al. 1999. Identical mutation in patients with limb girdle muscular dystrophy type 2B or Miyoshi myopathy suggests a role for modifier gene(s). Hum. Mol. Genet. 8: 871-887.
6. LocusLink Report (LocusID: 829). http://www.ncbi.nlm.nih.gov/LocusLink

CHROMOSOMAL LOCATION

Genetic locus: DYSF (human) mapping to 2p13.2; Dysf (mouse) mapping to 6 C3.

SOURCE

dysferlin (C-19) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of dysferlin 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-16635 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

dysferlin (C-19) is recommended for detection of dysferlin 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).

dysferlin (C-19) is also recommended for detection of dysferlin in additional species, including equine, canine and porcine.

Suitable for use as control antibody for dysferlin siRNA (h): sc-43739, dysferlin siRNA (m): sc-77329, dysferlin shRNA Plasmid (h): sc-43739-SH, dysferlin shRNA Plasmid (m): sc-77329-SH, dysferlin shRNA (h) Lentiviral Particles: sc-43739-V and dysferlin shRNA (m) Lentiviral Particles: sc-77329-V.

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.

SELECT PRODUCT CITATIONS

1. Ho, M., et al. 2004. Disruption of muscle membrane and phenotype divergence in two novel mouse models of dysferlin deficiency. Hum. Mol. Genet. 13: 1999-2010.
2. Guo, L.T., et al. 2010. Evaluation of commercial dysferlin antibodies on canine, mouse and human skeletal muscle. Neuromuscul. Disord. 20: 820-825.
3. Fuller, S.J., et al. 2012. A novel non-canonical mechanism of regulation of MST3 (mammalian Sterile20-related kinase 3). Biochem. J. 442: 595-610.

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.