SANTA CRUZ BIOTECHNOLOGY, INC.

dysferlin (C-11): sc-398905

SANTA CRUZ BIOTECHNOLOGY

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. Locus Link (LocusID: 829). http://www.ncbi.nlm.nih.gov/LocusLink/

CHROMOSOMAL LOCATION

Genetic locus: DYSF (human) mapping to 2p13.2.

SOURCE

dysferlin (C-11) is a mouse monoclonal antibody raised against amino acids 84-150 mapping near the N-terminus of dysferlin of human origin.

PRODUCT

Each vial contains 200 µg IgG_3 in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

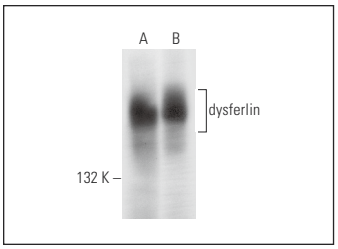
APPLICATIONS

dysferlin (C-11) is recommended for detection of dysferlin of human origin by Western Blotting (starting dilution 1:100, 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) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

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

Positive Controls: A-673 cell lysate: sc-2414 or human skeletal muscle extract: sc-363776.

DATA



dysferlin (C-11): sc-398905. Western blot analysis of dysferlin expression in A-673 whole cell lysate (**A**) and human skeletal muscle tissue extract (**B**).

RESEARCH USE

For research use only, not for use in diagnostic procedures.

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.