

Unveiling the degradative route of the V247M α-sarcoglycan mutant responsible for LGMD-2D.

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Hum Mol Genet. 2014 Jul 15;23(14):3746-58. doi: 10.1093/hmg/ddu088. Epub 2014 Feb 23.

[Rescue of sarcoglycan mutations by inhibition of endoplasmic reticulum quality control is associated with minimal structural modifications.](#)

Soheili T, Gicquel E, Poupiot J, N'Guyen L, Le Roy F, Bartoli M, Richard I.

Hum Mutat. 2012 Feb;33(2):429-39. doi: 10.1002/humu.21659. Epub 2011 Dec 22.

PMID: 22095924 [PubMed - indexed for MEDLINE]

[Nitric oxide donor and non steroidal anti inflammatory drugs as a therapy for muscular dystrophies: Evidence from a safety study with pilot efficacy measures in adult dystrophic patients.](#)

[D'Angelo MG](#) ,
[Boneschi F](#)
[Sciorati C](#)
[Bonato S](#)
[Brighina E](#)
[Comi GP](#)
[Turconi AC](#)
[Magri F](#)
[Stefanoni G](#)
[Brunelli S](#)
[Bresolin N](#)
[Cattaneo D](#)
[Clementi E](#)

[Gandossini S](#) ,

[Martinelli](#)

,

E. Medea Scientific Institute [Pier Bassi Pisa](#) 2012 Apr;65(4):472-9. Epub 2012 Jan 25.

[A dual acting compound releasing nitric oxide \(NO\) and ibuprofen, NCX 320, shows significant therapeutic effects in a mouse model of muscular dystrophy.](#)

Sciorati C, Miglietta D, Buono R, Pisa V, Cattaneo D, Azzoni E, Brunelli S, Clementi E.

Pharmacol Res. 2011 Sep;64(3):210-7. Epub 2011 May 12.

[Co-administration of ibuprofen and nitric oxide is an effective experimental therapy for muscular dystrophy, with immediate applicability to humans.](#)

Sciorati C, Buono R, Azzoni E, Casati S, Ciuffreda P, D'Angelo G, Cattaneo D, Brunelli S, Clementi E.

Br J Pharmacol. 2010 Jul;160(6):1550-60.

[Mannosidase I inhibition rescues the human alpha- **sarcoglycan** R77C recurrent mutation.](#)

Bartoli M, Gicquel E, Barrault L, Soheili T, Malissen M, Malissen B, Vincent-Lacaze N, Perez N, Udd B, et al. Mannosidase I inhibition rescues the human alpha-**sarcoglycan** R77C recurrent mutation. *Hum Mol Genet*. 2008 May 1;17(9):1214-21. doi: 10.1093/hmg/ddn029. Epub 2008 Feb 5.

PMID: 18252745 [PubMed - indexed for MEDLINE]