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[Arch Phys Med Rehabil.](#) 2017 Apr 30. pii: S0003-9993(17)30266-6.
doi: 10.1016/j.apmr.2017.04.004. [Epub ahead of print]

Six-Minute Walk Test as a Measure of Walking Capacity in Ambulatory Individuals With Amyotrophic Lateral Sclerosis.

[Sanjak M](#)¹, [Langford V](#)², [Holsten S](#)², [Rozario N](#)³, [Patterson CGM](#)³,
[Bravver E](#)⁴, [Bockenek WL](#)⁵, [Brooks BR](#)⁴.

OBJECTIVE: To determine the validity of the 6-minute walk test (6MWT) as an outcome measure to evaluate walking capacity in ambulatory patients with amyotrophic lateral sclerosis (ALS).

[Contemp Clin Trials.](#) 2017 Jul;58:34-39. doi:
10.1016/j.cct.2017.04.008. Epub 2017 Apr 24.

Developing standardized corticosteroid treatment for Duchenne muscular dystrophy.

[Guglieri M](#)¹, [Bushby K](#)², [McDermott MP](#)³, [Hart KA](#)³, [Tawil R](#)³, [Martens WB](#)³, [Herr BE](#)³, [McColl E](#)⁴, [Wilkinson J](#)⁴, [Kirschner J](#)⁵, [King WM](#)³, [Eagle M](#)², [Brown MW](#)³, [Willis T](#)⁶, [Hirtz D](#)⁷, [Shieh PB](#)⁸, [Straub V](#)², [Childs AM](#)⁹, [Ciafaloni E](#)³, [Butterfield RJ](#)¹⁰, [Horrocks I](#)¹¹, [Spinty S](#)¹², [Flanigan KM](#)¹³, [Kuntz NL](#)¹⁴, [Baranello G](#)¹⁵, [Roper H](#)¹⁶, [Morrison L](#)¹⁷, [Mah JK](#)¹⁸, [Manzur AY](#)¹⁹, [McDonald CM](#)²⁰, [Schara U](#)²¹, [von der Hagen M](#)²², [Barohn RJ](#)²³, [Campbell C](#)²⁴, [Darras BT](#)²⁵, [Finkel RS](#)²⁶, [Vita G](#)²⁷, [Hughes I](#)²⁸, [Mongini T](#)²⁹, [Pegoraro E](#)³⁰, [Wicklund M](#)³¹, [Wilichowski E](#)³², [Bryan Burnette W](#)³³, [Howard JF](#)³⁴, [McMillan HJ](#)³⁵, [Thangarajh M](#)³⁶, [Griggs RC](#)³.

Abstract

Despite corticosteroids being the only treatment documented to improve strength and function in boys with Duchenne muscular dystrophy (DMD) corticosteroid prescription is inconsistent and in some countries, corticosteroids are not prescribed. We are conducting a clinical trial that (1) compares the 3 most frequently prescribed corticosteroid regimens; (2) standardizes treatment of DMD complications; and (3) standardizes prevention of corticosteroid side effects. Investigators at 38 sites in 5 countries plan to recruit 300 boys aged 4-7 who are randomly assigned to one of three regimens: daily prednisone; daily deflazacort; or intermittent prednisone (10days on/10days off). Boys are followed for a minimum of 3years to assess the relative effectiveness and adverse event profiles of the different regimens. The primary outcome is a 3-dimensional variable consisting of log-transformed time to rise from the floor, forced vital capacity, and subject/parent satisfaction with treatment, each averaged over all post-baseline visits. The study protocol includes evidence- and consensus-based treatment of DMD complications and of corticosteroid side effects. This study seeks to establish a standard corticosteroid regimen for DMD. Since all new interventions for DMD are being developed as add-on therapies to corticosteroids, defining the optimum regimen is of importance for all new treatments.

[Muscle Nerve](#). 2017 Jun;55(6):922-927. doi: 10.1002/mus.25453. Epub 2017 Feb 12.

Episodic weakness and Charcot-marie-tooth disease due to a mitochondrial MT-ATP6 mutation.

[Panosyan FB](#)¹, [Tawil R](#)¹, [Herrmann DN](#)¹.

INTRODUCTION:

Episodic muscle weakness is the hallmark of a heterogeneous group of disorders known as periodic paralysis. A majority are due to single nucleotide mutations causing membrane depolarization.

[Clin Transplant](#). 2017 May;31(5). doi: 10.1111/ctr.12952. Epub 2017 Apr 17.

Relationship between pre-transplant physical function and outcomes after kidney transplant.

[Lorenz EC](#)^{1,2}, [Cheville AL](#)³, [Amer H](#)^{1,2}, [Kotajarvi BR](#)³, [Stegall MD](#)^{2,4}, [Petterson TM](#)⁵, [Kremers WK](#)^{2,5}, [Cosio FG](#)^{1,2}, [LeBrasseur NK](#)³.

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Abstract

BACKGROUND:

Performance-based measures of physical function predict morbidity following non-transplant surgery. Study objectives were to determine whether physical function predicts outcomes after kidney transplant and assess how physical function changes post-transplant.

[Disabil Rehabil.](#) 2017 Feb 7:1-8. doi: 10.1080/09638288.2017.1283712. [Epub ahead of print]

Patient reported outcomes in GNE myopathy: incorporating a valid assessment of physical function in a rare disease.

[Slota C](#)^{1,2}, [Bevans M](#)³, [Yang L](#)³, [Shrader J](#)⁴, [Joe G](#)⁴, [Carrillo N](#)^{1,5}.

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3c National Institutes of Health Clinical Center , Bethesda , MD , USA.

4d Rehabilitation Medicine Department , National Institutes of Health , Bethesda , MD , USA.

5e National Human Genome Research Institute, National Institutes of Health , Bethesda , MD , USA.

BACKGROUND:

The aim of this analysis was to evaluate the psychometric properties of three patient reported outcome (PRO) measures characterizing physical function in GNE myopathy: the Human Activity Profile, the Inclusion Body Myositis Functional Rating Scale, and the Activities-specific Balance Confidence scale.

[Muscle Nerve.](#) 2016 Feb;53(2):183-90. doi: 10.1002/mus.24725. Epub 2015 Dec 29.

<http://www.ncbi.nlm.nih.gov/pubmed/26044513>

Myotonic dystrophy health index: Correlations with clinical tests and patient function.

[Heatwole C](#)¹, [Bode R](#)², [Johnson NE](#)³, [Dekdebrun J](#)¹, [Dilek N](#)¹, [Eichinger K](#)¹, [Hilbert JE](#)¹, [Logigian E](#)¹, [Luebke E](#)¹, [Martens W](#)¹, [Mcdermott MP](#)^{1,4}, [Pandya S](#)¹, [Puwanant A](#)⁵, [Rothrock N](#)⁶, [Thornton C](#)¹, [Vickrey BG](#)^{7,8}, [Victorson D](#)⁶, [Moxley RT 3rd](#)¹.

[Neurology.](#) 2016 Nov 15;87(20):2123-2131. Epub 2016 Aug 26.

Efficacy and safety of deflazacort vs prednisone and placebo for Duchenne muscular dystrophy.

[Griggs RC](#)¹, [Miller JP](#)², [Greenberg CR](#)², [Fehlings DL](#)², [Pestronk A](#)², [Mendell JR](#)², [Moxley RT 3rd](#)², [King W](#)², [Kissel JT](#)², [Cwik V](#)², [Vanasse M](#)², [Florence JM](#)², [Pandya S](#)², [Dubow JS](#)², [Meyer JM](#)².

OBJECTIVE:

To assess safety and efficacy of deflazacort (DFZ) and prednisone (PRED) vs placebo in Duchenne muscular dystrophy (DMD).

[Neuromuscul Disord](#). 2015 Aug;25(8):625-31. doi:
10.1016/j.nmd.2015.04.013. Epub 2015 May 7.
<http://www.ncbi.nlm.nih.gov/pubmed/26022999>

Disease course and therapeutic approach in dermatomyositis: A four-center retrospective study of 100 patients.

[Johnson NE](#)¹, [Arnold WD](#)², [Hebert D](#)³, [Gwathmey K](#)⁴, [Dimachkie MM](#)⁵,
[Barohn RJ](#)⁵, [McVey AL](#)⁵, [Pasnoor M](#)⁵, [Amato AA](#)⁴, [McDermott MP](#)⁶, [Kissel J](#)², [Heatwole CR](#)⁷.

[Ann Clin Transl Neurol](#). 2015 Jul;2(7):739-47. doi: 10.1002/acn3.208.
Epub 2015 May 7. <http://www.ncbi.nlm.nih.gov/pubmed/26273686>

A randomized controlled trial of exercise in spinal and bulbar muscular atrophy.

[Shrader JA](#)¹, [Kats I](#)², [Kokkinis A](#)², [Zampieri C](#)¹, [Levy E](#)¹, [Joe GO](#)¹,
[Woolstenhulme JG](#)¹, [Drinkard BE](#)¹, [Smith MR](#)¹, [Ching W](#)¹, [Ghosh L](#)², [Fox D](#)²,
[Auh S](#)³, [Schindler AB](#)², [Fischbeck KH](#)², [Grunseich C](#)².

[Arthritis Care Res \(Hoboken\)](#). 2015 Jan;67(1):94-101. doi:
10.1002/acr.22468.
<http://www.ncbi.nlm.nih.gov/pubmed/25201017>

Lower extremity peak force and gait kinematics in individuals with inclusion body myositis.

[Davenport TE](#)¹, [Benson K](#), [Baker S](#), [Gracey C](#), [Rakocevic G](#), [McElroy B](#),
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[Neuromuscul Disord](#). 2014 Dec;24(12):1063-7. doi:
10.1016/j.nmd.2014.07.006. Epub 2014 Aug 7.
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Atypical presentation of GNE myopathy with asymmetric hand weakness.

[de Dios JK](#)¹, [Shrader JA](#)², [Joe GO](#)², [McClellan JC](#)³, [Williams K](#)², [Evers R](#)⁴,
[Malicdan MC](#)¹, [Ciccone C](#)¹, [Mankodi A](#)⁵, [Huizinga M](#)¹, [McKew JC](#)⁶,
[Bluemke DA](#)⁴, [Gahl WA](#)¹, [Carrillo-Carrasco N](#)⁷.

[Clin Exp Rheumatol](#). 2014 Sep-Oct;32(5):689-96. Epub 2014 Jul 28.
<http://www.ncbi.nlm.nih.gov/pubmed/25068290>

Novel assessment tools to evaluate clinical and laboratory responses in a subset of patients enrolled in the Rituximab in Myositis trial.

[Rider LG¹](#), [Yip AL](#), [Horkayne-Szakaly I](#), [Volochoyev R](#), [Shrader JA](#), [Turner ML](#), [Kong HH](#), [Jain MS](#), [Jansen AV](#), [Oddis CV](#), [Fleisher TA](#), [Miller FW](#)

[Brain](#). 2013 Jul;136(Pt 7):2189-200. doi: 10.1093/brain/awt133. Epub 2013 Jun 13. <http://www.ncbi.nlm.nih.gov/pubmed/23771340>

Non-dystrophic myotonia: prospective study of objective and patient reported outcomes.

[Trivedi JR¹](#), [Bundy B](#), [Statland J](#), [Salajegheh M](#), [Rayan DR](#), [Venance SL](#), [Wang Y](#), [Fialho D](#), [Matthews E](#), [Cleland J](#), [Gorham N](#), [Herbelin L](#), [Cannon S](#), [Amato A](#), [Griggs RC](#), [Hanna MG](#), [Barohn RJ](#); [CINCH Consortium](#).

[Muscle Nerve](#). 2012 Oct;46(4):482-9. doi: 10.1002/mus.23402.
<http://www.ncbi.nlm.nih.gov/pubmed/22987687>

A quantitative measure of handgrip myotonia in non-dystrophic myotonia.

[Statland JM¹](#), [Bundy BN](#), [Wang Y](#), [Trivedi JR](#), [Raja Rayan D](#), [Herbelin L](#), [Donlan M](#), [McLin R](#), [Eichinger KJ](#), [Findlater K](#), [Dewar L](#), [Pandya S](#), [Martens WB](#), [Venance SL](#), [Matthews E](#), [Amato AA](#), [Hanna MG](#), [Griggs RC](#), [Barohn RJ](#); [CINCH Consortium](#).

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Open-label trial of recombinant human insulin-like growth factor 1/recombinant human insulin-like growth factor binding protein 3 in myotonic dystrophy type 1.

[Heatwole CR¹](#), [Eichinger KJ](#), [Friedman DI](#), [Hilbert JE](#), [Jackson CE](#), [Logigian EL](#), [Martens WB](#), [McDermott MP](#), [Pandya SK](#), [Quinn C](#), [Smirnow AM](#), [Thornton CA](#), [Moxley RT 3rd](#).

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[**Efficacy and safety of dutasteride in patients with spinal and bulbar muscular atrophy: a randomised placebo-controlled trial.**](#)

[Fernández-Rhodes LE](#), [Kokkinis AD](#), [White MJ](#), [Watts CA](#), [Auh S](#), [Jeffries NO](#), [Shrader JA](#), [Lehky TJ](#), [Li L](#), [Ryder JE](#), [Levy EW](#), [Solomon BI](#), [Harris-Love MO](#), [La Pean A](#), [Schindler AB](#), [Chen C](#), [Di Prospero NA](#), [Fischbeck KH](#).

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[Open-label trial of recombinant human insulin-like growth factor 1/recombinant human insulin-like growth factor binding protein 3 in myotonic dystrophy type 1](#)

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[Change in natural history of Duchenne muscular dystrophy with long-term corticosteroid treatment: implications for management.](#)

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[Predicting maximal grip strength using hand circumference.](#)

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Institute of Neurology, University College of London, London WC1N 3BG, UK.

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[Reply: Comment on alemtuzumab and inclusion body myositis](#)

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Poster presentation; NIH Biomedical Summer Research Internship
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