MD-NET publication list

10. APRIL 2017 (MD-NET)

Towards a functional pathology of hereditary neuropathies

Acta Neuropathologica 2017 Apr

Weis J, Claeys KG, Roos A, Azzedine H, Katona I, Schroder JM, Senderek J.

6. MÄRZ 2017 (MD-NET)

Differential roles of α-, β-, and γ-actin in axon growth and collateral branch formation in motoneurons.

Moradi M, Sivadasan R, Saal L, Lüningschrör P, Dombert B, Rathod RJ, Dieterich DC, Blum R, Sendtner M.

J Cell Biol. 2017 Mar 6;216(3):793-814. doi: 10.1083/jcb.201604117. Epub 2017 Feb 28.

14. OKTOBER 2016 (MD-NET)

The Caveolin-3 G56S sequence variant of unknown significance: Muscle biopsy findings and functional cell biological analysis

Proteomics Clin Appl. 2016 Oct 14. doi: 10.1002/prca.201600007. [Epub ahead of print] Brauers E, Roos A, Kollipara L, Zahedi RP, Beckmann A, Mohanadas N, Bauer H, Häusler M, Thoma S, Kress W, Senderek J, Weis J.

8. OKTOBER 2015 (MD-NET)

Tumor Necrosis Factor Alpha and Insulin-Like Growth Factor 1 Induced Modifications of the Gene Expression Kinetics of Differentiating Skeletal Muscle Cells

Plos One

Meyer, SU; Krebs, S; Thirion, C; Blum, H; Krause, S; Pfaffl, MW

7. OKTOBER 2015 (MD-NET)

Long-term follow-up in patients with Congenital Cataract Facial Dysmorphism Neuropathy (CCFDN) Syndrome

Neurology 2014 Oct 7;83:1337-1344

Walter MC, Bernert G, Zimmermann U, Müllner-Eidenböck A, Moser E, Kalaydjieva L, Lochmüller H, Müller-Felber W.

13. AUGUST 2015 (MD-NET)

Integrative Analysis of MicroRNA and mRNA Data Reveals an Orchestrated Function of MicroRNAs in Skeletal Myocyte Differentiation in Response to TNF-α or IGF1

Plos One

Meyer, SU; Sass, S; Mueller, NS; Krebs, S; Bauersachs, S; Kaiser, S; Blum, H; Thirion, C; Krause, S; Theis, FJ; Pfaffl, MW

25. JULI 2015 (MD-NET)

50 years to diagnosis – autosomal dominant tubular aggregate myopathy caused by a novel STIM1 mutation. Neuromuscul Disord 2015

Neuromuscul Disord. 2015 Jul;25(7):577-84. doi: 10.1016/j.nmd.2015.04.005. Epub 2015 Apr 16.

Walter MC, Rossius M, Zitzelsberger M, Vorgerd M, Müller-Felber W, Ertl-Wagner B, Zhang Y, Brinkmeier H, Senderek J, Schoser B

25. MAI 2015 (MD-NET)

Transcriptional regulator PRDM12 is essential for human pain perception

Nat Genet. 2015 Jul;47(7):803-8. doi: 10.1038/ng.3308. Epub 2015 May 25

Chen YC, Auer-Grumbach M, Matsukawa S, Zitzelsberger M, Themistocleous AC, Strom TM, Samara C, Moore AW, Cho LT, Young GT, Weiss C, Schabhüttl M, Stucka R, Schmid AB, Parman Y, Graul-Neumann L, Heinritz W, Passarge E, Watson RM, Hertz JM, Moog U, Baumgartner M, Valente EM, Pereira D, Restrepo CM, Katona I, Dusl M, Stendel C, Wieland T, Stafford F, Reimann

F, von Au K, Finke C, Willems PJ, Nahorski MS, Shaikh SS, Carvalho OP, Nicholas AK, Karbani G, McAleer MA, Cilio MR, McHugh JC, Murphy SM, Irvine AD, Jensen UB, Windhager R, Weis J, Bergmann C, Rautenstrauss B, Baets J, De Jonghe P, Reilly MM, Kropatsch R, Kurth I, Chrast R, Michiue T, Bennett DL, Woods CG, Senderek J.

1. MAI 2015 (MD-NET)

Deciphering the impact of parameters influencing transgene expression kinetics after repeated cell transduction with integration-deficient retroviral vectors

Cytometry A. 2015 May;87(5):405-18. doi: 10.1002/cyto.a.22650.
Schott JW1, Jaeschke NM, Hoffmann D, Maetzig T, Ballmaier M, Godinho T, Cathomen T, Schambach A.

1. MAI 2015 (MD-NET)

ATOH8: a novel marker in human muscle fiber regeneration.

Histochem Cell Biol. 2015 May;143(5):443-52. doi: 10.1007/s00418-014-1299-6. Epub 2014 Dec 17.
Güttsches AK, Balakrishnan-Renuka A, Kley RA, Tegenthoff M, Brand-Saberi B, Vorgerd M.

25. FEBRUAR 2015 (MD-NET)

Abnormal proliferation and spontaneous differentiation of myoblasts from a symptomatic female carrier of X-linked Emery-Dreifuss muscular dystrophy

Neuromuscul Disord 2015; Feb 25:127-136
Meinke P, Schneiderat P, Srsen V, Korfali N, Thành PL, Wehnert M, Schirmer EC, Walter MC.

25. FEBRUAR 2015 (MD-NET)

Muscle ultrasound in classic infantile and adult Pompe disease: a useful screening tool in adults but not in infants.

Neuromuscul Disord. 2015 Feb;25(2):120-6. doi: 10.1016/j.nmd.2014.09.016. Epub 2014 Oct 22.
Vill K, Schessl J, Teusch V, Schroeder S, Blaschek A, Schoser B, Müller-Felber W.

25. FEBRUAR 2015 (MD-NET)

Abnormal proliferation and spontaneous differentiation of myoblasts from a symptomatic female carrier of X-linked Emery-Dreifuss muscular dystrophy

Neuromuscul Disord. 2015 Feb;25(2):127-36. doi: 10.1016/j.nmd.2014.09.012. Epub 2014 Oct 6.
Meinke P, Schneiderat P, Srsen V, Korfali N, Lê Thành P, Cowan GJ, Cavanagh DR, Wehnert M, Schirmer EC, Walter MC.

29. JANUAR 2015 (MD-NET)

TNF-α and IGF1 modify the microRNA signature in skeletal muscle cell differentiation

Cell Communication And Signaling
Meyer, SU; Thirion, C; Polesskaya, A; Bauersachs, S; Kaiser, S; Krause, S; Pfaffl, MW

18. DEZEMBER 2014 (MD-NET)

Comparative cost of illness analysis and assessment of health care burden of Duchenne and Becker muscular dystrophies in Germany.

Orphanet J Rare Dis. 2014 Dec 18;9(1):210.
Schreiber-Katz O, Klug C, Thiele S, Schorling E, Zowe J, Reilich P, Nagels KH, Walter MC.

7. OKTOBER 2014 (MD-NET)

Long-term follow-up in patients with CCFDN syndrome

Neurology. 2014 Oct 7;83(15):1337-44. doi: 10.1212/WNL.0000000000000874. Epub 2014 Sep 3.
Walter MC, Bernert G, Zimmermann U, Müllner-Eidenböck A, Moser E, Kalaydjieva L, Lochmüller H, Müller-Felber W.

1. OKTOBER 2014 (MD-NET)

Comparative economic impact of therapeutic innovation on health care burden of Duchenne

Muscular Dystrophy (DMD) using Becker Muscular Dystrophy (BMD) as a comparator for potential clinical outcome corridor.
Neuromuscular Disorders, Volume 24, Issues 9–10, October 2014, Pages 855, 19th International Congress of The World Muscle Society. Vorgestellt auf: 19th Int. Congress of the World Muscle Society, 07.-11.10.2014, Berlin, Germany.
Schreiber, O, Klug, C, Thiele, S, Schorling, E, Zowe, J, Reilich, P, Nagels, K, Walter, MC.

28. AUGUST 2014 (MD-NET)
High creatine kinase levels and white matter changes: clinical and genetic spectrum of congenital muscular dystrophies with laminin alpha-2 deficiency
Mol Cell Probes. 2014 Aug;28(4):118-22. doi: 10.1016/j.mcp.2013.11.002. Epub 2013 Nov 10.
Beytía Mde L, Dekomien G, Hoffjan S, Haug V, Anastasopoulos C, Kirschner J.

15. AUGUST 2014 (MD-NET)
Novel recessive myotilin mutation causes severe myofibrillar myopathy.
Neurogenetics. 2014 Aug;15(3):151-6. doi: 10.1007/s10048-014-0410-4. Epub 2014 Jun 14.
Schessl J, Bach E, Rost S, Feldkirchner S, Kubny C, Müller S, Hanisch FG, Kress W, Schoser B.

1. AUGUST 2014 (MD-NET)
Unusual multisystemic involvement and a novel BAG3 mutation revealed by NGS screening in a large cohort of myofibrillar myopathies.
Orphanet J Rare Dis. 2014 Aug 1;9:121. doi: 10.1186/s13023-014-0121-9.
Semmler AL, Sacconi S, Bach JE, Liebe C, Bürmann J, Kley RA, Ferbert A, Anderheiden R, Van den Bergh P, Martin JJ, De Jonghe P, Neuen-Jacob E, Müller O, Deschauer M, Bergmann M, Schröder JM, Vorgerd M, Schulz JB, Weis J, Kress W, Claeys KG.

2. JULI 2014 (MD-NET)
Use of whole-exome sequencing to determine the genetic basis of multiple mitochondrial respiratory chain complex deficiencies
JAMA. 2014 Jul 2;312(1):68-77. doi: 10.1001/jama.2014.7184.
Taylor RW, Pyle A, Griffin H, Blakely EL, Duff J, He L, Smertenko T, Alston CL, Neeve VC, Best A, Yarham JW, Kirschner J, Schara U, Talim B, Topaloglu H, Baric I, Holinski-Feder E, Abicht A, Czermin B, Kleinle S, Morris AA, Vassallo G, Gorman GS, Ramesh V, Turnbull DM, Santibanez-Koref M, McFarland R, Horvath R, Chinnery PF.

1. JUNI 2014 (MD-NET)
The impact of spinal muscular atrophies with regard to patients' quality of life: a socio-economic analysis of a German patient cohort
Clinical Neurophysiology, Volume 125, Supplement 1, June 2014, Page S196, ISSN 1388-2457, http://dx.doi.org/10.1016/S1388-2457(14)50639-6. Vorgestellt auf: 30th International Congress of Clinical Neurophysiology (ICCN) of the IFCN, March 20–23, 2014, Berlin, Germany.
Schreiber O, Klug C, Thiele S, Herrmann C, Zowe J, Reilich P, Nagels K, Walter MC.

1. JUNI 2014 (MD-NET)
Pregnancy and delivery in women with Pompe disease.
Mol Genet Metab. 2014 Jun;112(2):148-53. doi: 10.1016/j.ymgme.2014.03.010. Epub 2014 Mar 30.
Karabul N, Berndt J, Kornblum C, Kley RA, Wenninger S, Tiling N, Mengel E, Plöckinger U, Vorgerd M, Deschauer M, Schoser B, Hanisch F.

1. MAI 2014 (MD-NET)
Myopathy in Marinesco-Sjögren syndrome links endoplasmic reticulum chaperone dysfunction to nuclear envelope pathology
Acta Neuropathol. 2014 May;127(5):761-77. doi: 10.1007/s00401-013-1224-4. Epub 2013 Dec 21.

Roos A, Buchkremer S, Kollipara L, Labisch T, Gatz C, Zitzelsberger M, Brauers E, Nolte K, Schröder JM, Kirschner J, Jesse CM, Goebel HH, Goswami A, Zimmermann R, Zahedi RP, Senderek J, Weis J.

24. FEBRUAR 2014 (MD-NET)

Somatropin treatment of spinal muscular atrophy: a placebo-controlled, double-blind crossover pilot study

Neuromuscul Disord. 2014 Feb;24(2):134-42. doi: 10.1016/j.nmd.2013.10.011. Epub 2013 Nov 13.

Kirschner J, Schorling D, Hauschke D, Rensing-Zimmermann C, Wein U, Grieben U, Schottmann G, Schara U, Konrad K, Müller-Felber W, Thiele S, Wilichowski E, Hobbiebrunken E, Stettner GM, Korinthenberg R.

7. DEZEMBER 2013 (MD-NET)

Dominant-negative Effects of KCNQ2 Mutations are Associated with Epileptic Encephalopathy

Ann Neurol. 2013 Dec 7. doi: 10.1002/ana.24080. PMID: 24318194

Orhan G, Bock M, Schepers D, Ilina EI, Reichel SN, Löffler H, Jezutkovic N, Weckhuysen S, Mandelstam S, Suls A, Danker T, Guenther E, Scheffer IE, Jonghe PD, Lerche H, Maljevic S.

27. OKTOBER 2013 (MD-NET)

Mapping the differences in care for 5,000 spinal muscular atrophy patients, a survey of 24 national registries in North America, Australasia and Europe

J Neurol. 2014 Jan;261(1):152-63. doi: 10.1007/s00415-013-7154-1. Epub 2013 Oct 27.

Bladen CL, Thompson R, Jackson JM, Garland C, Wegel C, Ambrosini A, Pisano P, Walter MC, Schreiber O, Lusakowska A, Jedrzejowska M, Kostera-Pruszczyk A, van der Pol L, Wadman RI, Gredal O, Karaduman A, Topaloglu H, Yilmaz O, Matyushenko V, Rasic VM, Kosac A, Karcagi V, Garami M, Herczegfalvi A, Monges S, Moresco A, Chertkoff L, Chamova T, Guergueltcheva V, Butoianu N, Craiu D, Korngut L, Campbell C, Haberlova J, Strenkova J, Alejandro M, Jimenez A, Ortiz GG, Enriquez GV, Rodrigues M, Roxburgh R, Dawkins H, Youngs L, Lahdetie J, Angelkova N, Saugier-Veber P, Cuisset JM, Bloetzer C, Jeannet PY, Klein A, Nascimento A, Tizzano E, Salgado D, Mercuri E, Sejersen T, Kirschner J, Rafferty K, Straub V, Bushby K, Verschuuren J, Beroud C, Lochmüller H.

15. OKTOBER 2013 (MD-NET)

Identification of Xin-repeat proteins as novel ligands of the SH3 domains of nebulin and nebulette and analysis of their interaction during myofibril formation and remodeling

Molecular Biology Of The Cell

Eulitz, S; Sauer, F; Pelissier, MC; Boisguerin, P; Molt, S; Schuld, J; Orfanos, Z; Kley, RA; Volkmer, R; Wilmanns, M; Kirfel, G; van der Ven, PFM; Fürst, DO

16. SEPTEMBER 2013 (MD-NET)

Facioscapulohumeral muscular dystrophy and Charcot-Marie-Tooth neuropathy 1A-evidence for double trouble overlapping syndromes

Bmc Medical Genetics

Schreiber, O; Schneiderat, P; Kress, W; Rautenstrauss, B; Senderek, J; Schoser, B; Walter, MC

20. AUGUST 2013 (MD-NET)

Structural properties of EGCG-induced, nontoxic Alzheimer's disease Aβ oligomers.

J Mol Biol. 2012 Aug 24;421(4-5):517-24. doi: 10.1016/j.jmb.2012.01.013. Epub 2012 Jan 28.

Lopez del Amo JM, Fink U, Dasari M, Grelle G, Wanker EE,Bieschke J, Reif B.

11. JULI 2013 (MD-NET)

Mutations in RAB28, Encoding a Farnesylated Small GTPase, Are Associated with Autosomal-Recessive Cone-Rod Dystrophy.

Am J Hum Genet. 2013 Jul 11;93(1):110-7. doi: 10.1016/j.ajhg.2013.05.005. Epub 2013 Jun 6.

Roosing S, Rohrschneider K, Beryozkin A, Sharon D, Weisschuh N, Staller J, Kohl S, Zelinger L, Peters TA, Neveling K, Strom TM; European Retinal Disease Consortium, van den Born LI, Hoyng CB, Klaver CC, Roepman R, Wissinger B, Banin E, Cremers FP, den Hollander AI.

19. JUNI 2013 (MD-NET)

Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle.

Hum Mol Genet. 2013 Jun 19.

Klymiuk N, Blutke A, Graf A, Krause S, Burkhardt K, Wuensch A, Krebs S, Kessler B, Zakhartchenko V, Kurome M, Kemter E, Nagashima H, Schoser B, Herbach N, Blum H, Wanke R, Aartsma-Rus A, Thirion C, Lochmüller H, Walter MC, Wolf E.

19. JUNI 2013 (MD-NET)

Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle

Hum Mol Genet. 2013 Jun 19.

Klymiuk N, Blutke A, Graf A, Krause S, Burkhardt K, Wuensch A, Krebs S, Kessler B, Zakhartchenko V, Kurome M, Kemter E, Nagashima H, Schoser B, Herbach N, Blum H, Wanke R, Aartsma-Rus A, Thirion C, Lochmüller H, Walter MC, Wolf E.

30. MAI 2013 (MD-NET)

A novel optineurin truncating mutation and three glaucoma-associated missense variants in patients with familial amyothrophic lateral sclerosis in Germany

Neurobiol Aging. 2013 May;34(5):1516.e9-15. doi: 10.1016/j.neurobiolaging.2012.09.007. Epub 2012 Oct 10.

Weishaupt JH, Waibel S, Birve A, Volk AE, Mayer B, Meyer T, Ludolph AC, Andersen PM.

Source

Department of Neurology, Ulm University, Ulm, Germany. **jochen.weishaupt@uni-ulm.de**

23. MAI 2013 (MD-NET)

Proteomic characterization of aggregate components in an intrafamilial variable FHL1-associated myopathy.

Neuromuscul Disord. 2013 May;23(5):418-26. doi: 10.1016/j.nmd.2013.02.006. Epub 2013 Mar 13.

Feldkirchner S, Walter MC, Müller S, Kubny C, Krause S, Kress W, Hanisch FG, Schoser B, Schessl J.

18. APRIL 2013 (MD-NET)

ANO5 Gene Analysis in a Large Cohort of Patients with Anoctaminopathy: Confirmation of Male Prevalence and High Occurrence of the Common Exon 5 Gene Mutation.

Hum Mutat. 2013 Apr 18. doi: 10.1002/humu.22342.

Sarkozy A, Hicks D, Hudson J, Laval SH, Barresi R, Hilton-Jones D, Deschauer M, Harris E, Rufibach L, Hwang E, Bashir R, Walter MC, Krause S, van den Bergh P, Illa I, Pénisson-Besnier I, De Waele L, Turnbull D, Guglieri M, Schrank B, Schoser B, Seeger J, Schreiber H, Gläser D, Eagle M, Bailey G, Walters R, Longman C, Norwood F, Winer J, Muntoni F, Hanna M, Roberts M, Bindoff LA, Brierley C, Cooper RG, Cottrell DA, Davies NP, Gibson A, Gorman GS, Hammans S, Jackson AP, Khan A, Lane R, McConville J, McEntagart M, Al-Memar A, Nixon J, Panicker J, Parton M, Petty R, Price CJ, Rakowicz W, Ray P, Schapira AH, Swingler R, Turner C, Wagner KR, Maddison P, Shaw PJ, Straub V, Bushby K, Lochmüller H.

1. APRIL 2013 (MD-NET)

miR-411 is up-regulated in FSHD myoblasts and suppresses myogenic factors.

Orphanet J Rare Dis. 2013 Apr 5;8:55. doi: 10.1186/1750-1172-8-55.

Harafuji N, Schneiderat P, Walter MC, Chen YW.

14. FEBRUAR 2013 (MD-NET)

Treatment of dysferlinopathy with deflazacort: a double-blind, placebo-controlled clinical trial.
Orphanet J Rare Dis. 2013 Feb 14;8:26. doi: 10.1186/1750-1172-8-26.
Walter MC, Reilich P, Thiele S, Schessl J, Schreiber H, Reiners K, Kress W, Müller-Reible C, Vorgerd M, Urban P, Schrank B, Deschauer M, Schlotter-Weigel B, Kohnen R, Lochmüller H.

1. FEBRUAR 2013 (MD-NET)

Myopodin is an F-actin bundling protein with multiple independent actin-binding regions.
J Muscle Res Cell Motil. 2013 Feb;34(1):61-9. doi: 10.1007/s10974-012-9334-5. Epub 2012 Dec 9.
Linnemann A, Vakeel P, Bezerra E, Orfanos Z, Djinović-Carugo K, van der Ven PF, Kirfel G, Fürst DO.

16. JANUAR 2013 (MD-NET)

A new web-based method for automated analysis of muscle histology.
BMC Musculoskelet Disord. 2013 Jan 16;14:26. doi: 10.1186/1471-2474-14-26.
Pertl C, Eblenkamp M, Pertl A, Pfeifer S, Wintermantel E, Lochmüller H, Walter MC, Krause S, Thirion C.

1. JANUAR 2013 (MD-NET)

Filamin C-related myopathies: pathology and mechanisms
Acta Neuropathologica
Fürst, DO; Goldfarb, LG; Kley, RA; Vorgerd, M; Olivé, M; van der Ven, PFM

1. DEZEMBER 2012 (MD-NET)

Patient-specific protein aggregates in myofibrillar myopathies: laser microdissection and differential proteomics for identification of plaque components.
Proteomics. 2012 Dec;12(23-24):3598-609. doi: 10.1002/pmic.201100559. Epub 2012 Nov 5.
Feldkirchner S, Schessl J, Müller S, Schoser B, Hanisch FG.

1. NOVEMBER 2012 (MD-NET)

Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations.
J Neurol (2012) 259:838–850. DOI 10.1007/s00415-011-6262-z. Epub 2011 Oct 6.

Guergueltcheva V, Müller JS, Dusl M, Senderek J, Oldfors A, Lindbergh C, Maxwell S, Colomer J, Mallebrera CJ, Nascimento A, Vilchez JJ, Muelas N, Kirschner J, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Schlotter B, Schoser B, Herrmann R, Voit T, Steinlein OK, Najafi A, Urtizberea A, Soler DM, Muntoni F, Hanna MG, Chaouch A, Straub V, Bushby K, Palace J, Beeson D, Abicht A, Lochmüller H.

31. OKTOBER 2012 (MD-NET)

A combined laser microdissection and mass spectrometry approach reveals new disease relevant proteins accumulating in aggregates of filaminopathy patients.
Kley RA, Maerkens A, Leber Y, Theiss V, Schreiner A, van der Ven PF, Uszkoreit J, Stephan C, Eulitz S, Euler N, Kirschner J, Mueller K, Meyer HE, Tegenthoff M, Fuerst DO, Vorgerd M, Mueller T, Marcus K.
Klymiuk N, Thirion C, Burkhardt K, Wuensch A, Krause S, Krebs S, Graf A, Kessler B, Zakhartchenko V, Kurome M, Nagashima H, Schoser B, Herbach N, Blitke A, Blum H, Wanke R, Lochmüller H, Walter MC, Wolf E. Targeted deletion of DMD exon 52 in the pig results in biochemical and clinical hallmarks of Duchenne muscular dystrophy. Sci Trans Med 2011 submitted. Mol Cell Proteomics. 2012 Oct 31. [Epub ahead of print]

1. OKTOBER 2012 (MD-NET)

Muscle MRI findings in limb girdle muscular dystrophy type 2L
Sarkozy A, Deschauer M, Schrank B, Seeger J, Walter MC, Schoser B, Reilich P, Radunovic A, Schreiber H, Vaidya SS, Gläser D, Bushby K, Lochmüller H, Straub V.
Neuromuscul Disord. 2012 Oct 1;22 Suppl 2:S122-9. doi: 10.1016/j.nmd.2012.05.012.

1. SEPTEMBER 2012 (MD-NET)

Pathophysiology of protein aggregation and extended phenotyping in filaminopathy
Brain
Kley, RA; Serdaroglu-Oflazer, P; Leber, Y; Odgerel, Z; van der Ven, PFM; Olivé, M; Ferrer, I; Onipe, A; Mihaylov, M; Bilbao, JM; Lee, HS; Höhfeld, J; Djinovic-Carugo, K; Kong, K; Tegenthoff, M; Peters, SA; Stenzel, W; Vorgerd, M; Goldfarb, LG; Fürst, DO

1. AUGUST 2012 (MD-NET)

LINC complex alterations in DMD and EDMD/CMT fibroblasts.
Eur J Cell Biol. 2012 Aug;91(8):614-28. doi: 10.1016/j.ejcb.2012.03.003. Epub 2012 May 1.
Taranum S, Vaylann E, Meinke P, Abraham S, Yang L, Neumann S, Karakesisoglou I, Wehnert M, Noegel AA.

27. JUNI 2012 (MD-NET)

Congenital myasthenic syndromes: impact of gentotype-phentotype correlation on strategy and efficiency of genetic testing.
Abicht A, Dusl M, Gallenmüller C, Guergueltcheva C, Schara U, Marina AD, Almaras S, von der Hagen M, Huebner A, Chaouch A, Mihaylova M, Müller JS, Lochmüller H.
Hum Mutat. 2012 Oct;33(10):1474-84. doi: 10.1002/humu.22130. Epub 2012 Jun 27

18. JUNI 2012 (MD-NET)

Profound Effect of Profiling Platform and Normalization Strategy on Detection of Differentially Expressed MicroRNAs – A Comparative Study
Plos One
Meyer, SU; Kaiser, S; Wagner, C; Thirion, C; Pfaffl, MW

1. MAI 2012 (MD-NET)

Novel ANO5 mutations causing hyper-CK-emia, limb girdle muscular weakness and Miyoshi type of muscular dystrophy.
Muscle Nerve. 2012 May;45(5):740-2. doi: 10.1002/mus.23281.
Schessl J, Kress W, Schoser B.

29. MÄRZ 2012 (MD-NET)

A role for PLC1 in myotonic dystrophies type 1 and 2.
The FASEB Journal article fj.11-200337. Published online March 29, 2012.
Irene Faenza, William Blalock, Alberto Bavelloni, Benedikt Shoser, Roberta Fiume, Stephaniè Pacella, Manuela Piazzi, Antonietta D'Angelo, and Lucio Cocco.

23. MÄRZ 2012 (MD-NET)

Biomechanical characterization of a desminopathy in primary human myoblasts.
Biochem Biophys Res Commun. 2012 Mar 23;419(4):703-7. doi: 10.1016/j.bbrc.2012.02.083. Epub 2012 Feb 21.
Bonakdar N, Luczak J, Lautscham L, Czonstke M, Koch TM, Mainka A, Jungbauer T, Goldmann WH, Schröder R, Fabry B.

15. FEBRUAR 2012 (MD-NET)

Toward deconstructing the phenotype of late-onset Pompe disease.
Am J Med Genet C Semin Med Genet. 2012 Feb 15;160(1):80-8. doi: 10.1002/ajmg.c.31322. Epub 2012 Jan 17.
Schüller A, Wenninger S, Strigl-Pill N, Schoser B.

1. JANUAR 2012 (MD-NET)

Diagnose und Therapie der Muskeldystrophie Duchenne.
Monatsschr Kinderheilkd 2012 • 160:177–186. DOI 10.1007/s00112-011-2603-3.
J. Vry, U. Schara, S. Lutz, J. Kirschner

1. JANUAR 2012 (MD-NET)

Aktuelle Aspekte zur Klinik, Diagnose und Therapie von Muskelerkrankungen.
Neurol Rehabil 2011; 18 (1): 3 – 18, Neurologie & Rehabilitation Ausgabe 1-2012.
Schüller, B. Schoser

20. DEZEMBER 2011 (MD-NET)

A rat model of Charcot Marie Tooth disease 1A recapitulates disease variability and supplies biomarkers of axonal loss in patients
Fledrich F, Schlotter-Weigel B, Schnizer T, Wichert S, Stassart RM, Meyer zu Horste G, Weiss BG, Haag U, Walter MC, Rautenstrauss B, Paulus W, Nave KA, Rossner M, Sereda MW. Brain. 2012 Jan;135(Pt 1):72-87. doi: 10.1093/brain/awr322. Epub 2011 Dec 20.

1. DEZEMBER 2011 (MD-NET)

Reducing body myopathy and other FHL1 related muscular disorders
Schessl J, Feldkirchner S, Kubny C, Schoser B. Semin Pediatr Neurol. 2011 Dec;18(4):257-63. doi: 10.1016/j.spen.2011.10.007

30. NOVEMBER 2011 (MD-NET)

Distal myopathy with upper limb predominance caused by filamin C
haploinsufficiencGuergueltcheva, V., K. Peeters, J. Baets, C. Ceuterick-de Groote, J.-J. R Martin, A. Suls, E. De Vriendt, V. Mihaylova, T. Chamova, L. Almeida-Souza, E. Ydens, C. Tzekov, G. Hadjidekov, M. Gospodinova, K. Storm, E. Reyniers, S. Bichev, P. F. M. van der Ven, D. O. Fürst, V. Mitev, H.
Lochmüller, V. Timmerman, I. Tournev, P. De Jonghe und A. Jordanova (2011).
y. Neurology. 2011 Dec 13;77(24):2105-14. doi: 10.1212/WNL.0b013e31823dc51e. Epub 2011 Nov 30.

29. OKTOBER 2011 (MD-NET)

Adult-onset cerebellar ataxia due to mutations in CABC1/ADCK3
Horvath R, Czermin B, Gulati S, Demuth S, Houge G, Pyle A, Dineiger C, Blakely EL, Hassani A, Foley C, Brodhun M, Storm K, Kirschner J, Gorman GS, Lochmüller H, Holinski-Feder E, Taylor RW, Chinnery PF.. J Neurol Neurosurg Psychiatry. 2011 Oct 29.

6. OKTOBER 2011 (MD-NET)

Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations.
Guergueltcheva V, Dusl M, Senderek J, Oldfors A, Lindbergh C, Maxwell S, Colomer J, Mallebrera CJ, Nascimento A, Vilchez JJ, Muelas N, Kirschner J, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Schlotter B, Schoser B, Herrmann R, Voit T, Steinlein OK, Urtizberea A, Chaouch A, Straub V, Bushby K, Palace J, Beeson D, Abicht A, Lochmüller H, Müller JS. J Neurol 2011 Oct 6. PubMed PMID: 21975507.

1. OKTOBER 2011 (MD-NET)

Phenotypic heterogeneity in British patients with a founder mutation in the FHL1 gene
European Journal Of Human Genetics
Sarkozy, A; Windpassinger, C; Hudson, J; Dougan, CF; Lecky, B; Hilton-Jones, D; Eagle, M; Charlton, R; Barresi, R; Lochmüller, H; Bushby, K; Straub, V

7. AUGUST 2011 (MD-NET)

A retrospective clinical study of the treatment of slow-channel congenital myasthenic syndrome
Chaouch A, Müller JS, Guergueltcheva V, Dusl M, Schara U, Rakocević-Stojanović V, Lindberg C, Scola RH, Werneck LC, Colomer J, Nascimento A, Vilchez JJ, Muelas N, Argov Z, Abicht A, Lochmüller H. J Neurol 2011 Aug 7. [Epub ahead of print]

1. AUGUST 2011 (MD-NET)

A novel mutation in the myotilin gene (MYOT) causes a severe form of limb girdle muscular dystrophy 1A (LGMD1A)

Reilich P, Krause S, Schramm N, Klutzny U, Bulst S, Zehetmayer B, Schneiderat P, Walter MC, Schoser B, Lochmüller H. J Neurol 2011 Aug;258(8):1437-44.

1. AUGUST 2011 (MD-NET)

Nemaline myopathy caused by mutations in the nebulin gene may present as a distal myopathy.

Lehtokari VL, Pelin K, Herczegfalvi A, Karcagi V, Pouget J, Franques J, Pellissier JF, Figarella-Branger D, von der Hagen M, Huebner A, Schoser B, Lochmüller H, Wallgren-Pettersson C. Neuromuscul Disord 2011 Aug;21(8):556-62.

1. JULI 2011 (MD-NET)

Diabetes and neurodegeneration in Wolfram syndrome: a multicenter study of phenotype and genotype

Rohayem J, Ehlers C, Wiedemann B, Holl R, Oexle K, Kordonouri O, Salzano G, Meissner T, Burger W, Schober E, Huebner A, Lee-Kirsch MA; Wolfram Syndrome Diabetes Writing Group. Diabetes Care. 2011 Jul;34(7):1503-10.

10. JUNI 2011 (MD-NET)

Mutations in the N-terminal actin-binding domain of filamin C cause a distal myopathy

Duff RM, Tay V, Hackman P, Ravenscroft G, McLean C, Kennedy P, Steinbach A, Schöffler W, van der Ven PF, Fürst DO, Song J, Djinović-Carugo K, Penttilä S, Raheem O, Reardon K, Malandrini A, Gambelli S, Villanova M, Nowak KJ, Williams DR, Landers JE, Brown RH Jr, Udd B, Laing NG. Am J Hum Genet. 2011 Jun 10;88(6):729-40.

1. JUNI 2011 (MD-NET)

Recombination mapping of the susceptibility region for sporadic inclusion body myositis within the major histocompatibility complex

Scott AP, Laing NG, Mastaglia F, Needham M, Walter MC, Dalakas MC, Allcock RJ. J Neuroimmunol 2011 Jun;235(1-2):77-83.

1. JUNI 2011 (MD-NET)

New ophthalmic features in a family with triple A syndrome

Moschos MM, Margetis I, Koehler K, Gatzioufas Z, Huebner A. Int Ophthalmol. 2011 Jun;31(3):239-43.

1. JUNI 2011 (MD-NET)

Misregulated alternative splicing of BIN1 is associated with T tubule alterations and muscle weakness in myotonic dystrophy.

Fugier C, Klein AF, Hammer C, Vassilopoulos S, Ivarsson Y, Toussaint A, Tosch V, Vignaud A, Ferry A, Messaddeq N, Kokunai Y, Tsuburaya R, de la Grange P, Dembele D, Francois V, Precigout G, Boulade-Ladame C, Hummel MC, de Munain AL, Sergeant N, Laquerrière A, Thibault C, Deryckere F, Auboeuf D, Garcia L, Zimmermann P, Udd B, Schoser B, Takahashi MP, Nishino I, Bassez G, Laporte J, Furling D, Charlet-Berguerand N.
Nat Med 2011 Jun;17(6):720-5.

5. MAI 2011 (MD-NET)

The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene

Reilich P, Horvath R, Krause S, Schramm N, Turnbull DM, Trenell M, Hollingsworth KG, Gorman GS, Hans VH, Reimann J, Macmillan A, Turner L, Schollen A, Witte G, Czermin B, Holinski-Feder E, Walter MC, Schoser B, Lochmüller H. J Neurol 2011 May 5.

1. MAI 2011 (MD-NET)

Progressive external ophthalmoplegia as initial manifestation of sporadic late-onset nemaline myopathy

Wengert O, Meisel A, Kress W, Dekomien G, Angstwurm K, Heppner FL, Goebel HH, Stenzel W. J Neurol 2011 May;258(5):915-7.

1. MAI 2011 (MD-NET)

Long-term efficiency of intravenously administered immunoglobulin in anti-Yo syndrome with paraneoplastic cerebellar degeneration

Schessl J, Schuberth M, Reilich P, Schneiderat P, Strigl-Pill N, Walter MC, Schlotter-Weigel B, Schoser B. J Neurol 2011 May;258(5):946-7.

1. MÄRZ 2011 (MD-NET)

Alterations of excitation-contraction coupling and excitation coupled Ca(2+) entry in human myotubes carrying CAV3 mutations linked to rippling muscle.

Ullrich ND, Fischer D, Kornblum C, Walter MC, Niggli E, Zorzato F, Treves S. Hum Mutat. 2011 Mar;32(3):309-17.

1. MÄRZ 2011 (MD-NET)

Two siblings with triple A syndrome and novel mutation presenting as hereditary polyneuropathy

Dumić M, Barišić N, Rojnić-Putarek N, Kušec V, Stanimirović A, Koehler K, Huebner A. Eur J Pediatr. 2011 Mar;170(3):393-6.

11. FEBRUAR 2011 (MD-NET)

Hexosamine biosynthetic pathway mutations cause neuromuscular transmission defect

Senderek J, Müller JS, Dusl M, Strom TM, Guergueltcheva V, Diepolder I, Laval SH, Maxwell S, Cossins J, Krause S, Muelas N, Vilchez JJ, Colomer J, Mallebrera CJ, Nascimento A, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Steinlein OK, Schlotter B, Schoser B, Kirschner J, Herrmann R, Voit T, Oldfors A, Lindbergh C, Urtizberea A, von der Hagen M, Hübner A, Palace J, Bushby K, Straub V, Beeson D, Abicht A, Lochmüller H. Am J Hum Genet 2011 Feb 11;88(2):162-72.

1. FEBRUAR 2011 (MD-NET)

Acute liver failure with subsequent cirrhosis as the primary manifestation of TRMU mutations

Schara U, von Kleist-Retzow JC, Lainka E, Gerner P, Pyle A, Smith PM, Lochmüller H, Czermin B, Abicht A, Holinski-Feder E, Horvath R. J Inherit Metab Dis 2011 Feb;34(1):197-201.

1. FEBRUAR 2011 (MD-NET)

Genomic integration of adenoviral gene transfer vectors following transduction of fertilized mouse oocytes

Larochelle N, Stucka R, Rieger N, Schermelleh L, Schiedner G, Kochanek S, Wolf E, Lochmüller H. Transgenic Res 2011 Feb;20(1):123-35.

4. JANUAR 2011 (MD-NET)

Non-ATGinitiated translation directed by microsatellite expansions

Zu T, Gibbens B, Doty NS, Gomes-Pereira M, Huguet A, Stone MD, Margolis J, Peterson M, Markowski TW, Ingram MA, Nan Z, Forster C, Low WC, Schoser B, Somia NV, Clark HB, Schmechel S, Bitterman PB, Gourdon G, Swanson MS, Moseley M, Ranum LP.
Proc Natl Acad Sci U S A 2011 Jan 4;108(1):260-5.

1. JANUAR 2011 (MD-NET)

Sarcoglycanopathies

Kirschner J, Lochmüller H. Handb Clin Neurol 2011;101:41-6.

1. JANUAR 2011 (MD-NET)

Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency

Kemp JP, Smith PM, Pyle A, Neeve VC, Tuppen HA, Schara U, Talim B, Topaloglu H, Holinski-Feder E, Abicht A, Czermin B, Lochmüller H, McFarland R, Chinnery PF, Chrzanowska-Lightowlers ZM, Lightowlers RN, Taylor RW, Horvath R. Brain 2011 Jan;134(Pt 1):183-95.

1. JANUAR 2011 (MD-NET)

A founder mutation in anoctamin 5 is a major cause of limb girdle muscular dystrophy

Hicks D, Sarkozy A, Muelas N, Koehler K, Huebner A, Hudson G, Chinnery PF, Barresi R, Eagle M, Polvikoski T, Bailey G, Miller J, Radunovic A, Hughes PJ, Roberts R, Krause S, Walter MC, Laval SH, Straub V, Lochmüller H, Bushby K. Brain 2011;134:171-182.

1. JULI 2010 (MD-NET)

Facioscapulohumeral muscular dystrophy presenting with unusual phenotypes and atypical morphological features of vacuolar myopathy

Journal Of Neurology

Reilich, P; Schramm, N; Schoser, B; Schneiderat, P; Strigl-Pill, N; Müller-Höcker, J; Kress, W; Ferbert, A; Rudnik-Schöneborn, S; Noth, J; Lochmüller, H; Weis, J; Walter, MC

1. APRIL 2010 (MD-NET)

The p.G154S mutation of the alpha-B crystallin gene (CRYAB) causes late-onset distal myopathy

Neuromuscular Disorders

Reilich, P; Schoser, B; Schramm, N; Krause, S; Schessl, J; Kress, W; Müller-Höcker, J; Walter, MC; Lochmuller, H

1. APRIL 2010 (MD-NET)

Divergent Molecular Effects of Desmin Mutations on Protein Assembly in Myofibrillar Myopathy

Journal Of Neuropathology And Experimental Neurology

Levin, J; Bulst, S; Thirion, C; Schmidt, F; Bötzel, K; Krause, S; Pertl, C; Kretzschmar, H; Walter, MC; Giese, A; Lochmüller, H