

Publication list CMT-NET

Targeting PI3K/Akt/mTOR signaling in rodent models of PMP22 gene-dosage diseases

EMBO Mol Med. 2024. >>[PubMed-Link](#)<<

Doris Krauter, Daniela Stausberg, Timon J Hartmann, Stefan Volkmann, Theresa Kungl, David A Rasche, Gesine Saher, Robert Fledrich, Ruth M Stassart, Sandra Goebbels, David Ewers, Michael W Sereda

A polymorphic AT-repeat causes frequent allele dropout for an MME mutational hotspot exon

J Med Genet. 2022 Mar 22. Online ahead of print >>[PubMed-Link](#)<<

Helle Høyer, Hilde T Hilmarsen, Raute Sunder-Plassmann, Geir J Braathen, Peter M Andersen, Christian Beetz, Sandra Hacker, Øystein L Holla, Ingo Kurth, Wolfgang N Löscher, Simone B C F Reiter, Sabine Rudnik-Schöneborn, Linda Strand, Reinhard Windhager, Martina Witsch-Baumgartner, Jan Senderek, Michaela Auer-Grumbach

Neuromuscular rehabilitation – what to do?

Review Curr Opin Neurol. 2021 Oct 1. >>[PubMed-Link](#)<<

Melissa R Mandarakas, Peter Young, Joshua Burns

Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia

Brain. 2021 Jun 22. >>[PubMed-Link](#)<<

Manuela Wiessner et al.

The genetic landscape of axonal neuropathies in the middle-aged and elderly Focus on MME

Neurology 2020 Dec 15;95(24):e3163-e3179. Epub 2020 Nov 3. >>[PubMed-Link](#)<<

Senderek J, Lassuthova P, Kabzinska D, Abreu L, Baets J, Beetz C, Braathen GJ, Brenner D, Dalton J, Dankwa L, Deconinck T, De Jonghe P, Drager B, Eggermann K, Ellis M, Fischer C, Stojkovic T, Herrmann DN, Horvath R, Hoyer H, Iglseider S, Kennerson M, Kinslechner K, Kohler JN, Kurth I, Laing NG, Lamont PJ, Loscher WN, Ludolph A, Marques W, Nicholson G, Ong R, Petri S, Ravenscroft G, Rebelo A, Ricci G, Rudnik-Schöneborn S, Schirmacher A, Schlotter-Weigel B, Schoels L, Schule R, Synofzik M, Francou B, Strom TM, Wagner J, Walk D, Wanschitz J, Weinmann D, Weishaupt J, Wiessner M, Windhager R, Young P, Zuchner S, Toegel S, Seeman P, Kochanski A, Auer-Grumbach M

Demyelinating Charcot-Marie-Tooth neuropathy associated with FBLN5 mutations

European Journal of Neurology 2020; 27(12): 2568-2574; >>[PubMed-Link](#)<<

Brozkova DS, Stojkovic T, Haberlova J, Mazanec R, Windhager R, Rosenegger PF, Hacker S, Zuchner S, Kochanski A, Leonard-Louis S, Francou B, Latour P, Senderek J, Seeman P, Auer-Grumbach M.

Charcot-Marie-Tooth disease and hereditary motor neuropathies – Update 2020

Medizinische Genetik 2020; 32(3): 207-219; >>[MedGen-Link](#)<<

Rudnik-Schöneborn S, Auer-Grumbach M, Senderek J

Pregnancy outcome in Charcot-Marie-Tooth disease: results of the CMT-NET cohort study in Germany

European Journal of Neurology 2020; 27(8): 1390-1396; >>[PubMed-Link](#)<<

Rudnik-Schöneborn S, Thiele S, Walter MC, Reinecke L, Sereda M, Schöneborn R, Elbracht M.

Axo-glial interaction in the injured PNS

Developmental Neurobiology: 2020 Jul >>[PubMed-Link](#)<<

Stassart RM, Woodhoo A.

Intraepidermal nerve fibre density as biomarker in Charcot-Marie-Tooth disease type 1A

Brain Communications 2020; 2(1): 14 >>[PubMed-Link](#)<<

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Axo-glia interdependence in peripheral nerve development

Development 2019 Dec 12; 146(21): 12

Fledrich R, Kungl T, Nave KA, Stassart RM

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Neuromuscul Disord. 2019 May;29(5):392-397.

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Nat Commun. 2019 Apr 1;10(1):1467

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DETAILS

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Neurology 2019 Mar 27

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DETAILS

26. MÄRZ 2019 (CMT-NET)

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Cell Reports 2019; 26(13): 3484; 2019 Mar 26

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European Journal of Paediatric Neurology 2019; 23(2): 254-261; Epub 2018 Dec 29

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Early short-term PXT3003 combinational therapy delays disease onset in a transgenic rat model of Charcot-Marie-Tooth disease 1A (CMT1A)

PLoS One. 2019 Jan 16;14(1):e0209752. doi: 10.1371/journal.pone.0209752. eCollection 2019.

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DETAILS

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Neurobiol Dis. 2018 Nov 20;124:218-229. doi: 10.1016/j.nbd.2018.11.019

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DETAILS

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Rare Variants in MME, Encoding Metalloprotease Nephrilysin, Are Linked to Late-Onset Autosomal-Dominant Axonal Polyneuropathies

The American Journal of Human Genetics 99, Sept 16

Michaela Auer-Grumbach, Stefan Toegel, Maria Schabhüttl, Daniela Weinmann, Catharina Chiari, David L.H. Bennett, Christian Beetz, Dennis Klein, Peter M. Andersen, Ilka Böhme, Regina Fink-Puches, Michael Gonzalez, Matthew B. Harms, William Motley, Mary M. Reilly, Wilfried Renner, Sabine Rudnik-Schöneborn, Beate Schlotter-Weigel, Andreas C. Themistocleous, Jochen H. Weishaupt, Albert C. Ludolph, Thomas Wieland, Feifei Tao, Lisa Abreu, Reinhard Windhager, Manuela Zitzelsberger, Tim M. Strom, Thomas Walther, Steven S. Scherer, Stephan Züchner, Rudolf Martini, and Jan Senderek