2026

Phenotype and Genetics of Spinocerebellar Ataxia Type 27B: Novel Movement-disorder Features, Cognitive Impairment, and Repeat Expansion Findings.
Rashedi R, Peemöller F, Erdmann H, Gelderblom M, Hidding U, Ganos C, Chen R, Abicht A, Zittel S.
https://pubmed.ncbi.nlm.nih.gov/42371259/

Bridging past and future: the evolution of genetic diagnosis in FSHD and the role of emerging technologies in a globalized framework.
Strafella C, Erdmann H, Bevilacqua JA, Ricci E, Ravenscroft G, Matsumura T, Vishnu VY, Sherif RE, Oflazer P, Muratori D, Figueiredo F, Sansone V, Sacconi S, Ricci G, Filosto M, Evangelista T, Rosa AL, Belayew A, Abicht A, Magdinier F, Giardina E.
https://pubmed.ncbi.nlm.nih.gov/41996965/

The p.(Leu97Ile) variant expands the genetic landscape of NEFL-associated Charcot-Marie-tooth neuropathies.
Oeztuerk M, Walli S, Muhmann D, Choueiri C, Dobelmann V, Abicht A, Leube B, Schara-Schmidt U, Meuth SG, Horvath R, Lochmueller H, Roos A, Ruck T.
https://pubmed.ncbi.nlm.nih.gov/42017539/

Atypical Clinical Course of Griscelli Syndrome Type 2 With Primarily Neurologic Presentation and Adult-Onset in a 46-Year-Old Male.
Papingi D, Kutsche M, Lichtenfeld H, Kortüm F, Abicht A, Herrmann L, Herget T.
https://pubmed.ncbi.nlm.nih.gov/41851022/

Repeat-associated ataxias in a German patient cohort analysed by targeted parallel long-read sequencing.
Erdmann H, Schaub A, Lucas MC, Scholz V, Benet-Pages A, Becker K, Dineiger C, Mayer V, van Buren I, Breithausen E, Akbari K, Cordts I, Sauer M, Schneider C, Krakowsky R, Schnabel F, Dunker K, Fabritius L, Gerb J, Grabova D, Möhwald K, Näher M, Steinmetz K, Thiessen F, Jäck A, Schneider-Gold C, Zittel S, Petersen C, Schreyer I, Mämecke L, Wilfling S, Wunderlich G, Brenner D, Hellenbroich Y, Muhle K, Huchtemann T, Claus I, Klopstock T, Strupp M, Levin J, Höglinger G, Huppert D, Becker-Bense S, Filippopulos F, Kilpert F, Leitão E, Kaya S, Depienne C, Schöberl F, Neuhann T, Holinski-Feder E, Zwergal A, Abicht A.
https://pubmed.ncbi.nlm.nih.gov/40898875/

Mitochondrial energetic failure underlies FLVCR1-related sensory neuropathy.
Bertino F, Zanin Venturini DI, Grasso E, Kopecka J, Salio C, Gnutti B, Basnet RM, Bellini S, Mignani L, Zhao B, Kleefeld F, Hentschel A, Magnani F, Fiorito V, Abalai RE, Metani L, Allocco AL, Petrillo S, De Giorgio F, Ammirata G, Salsano E, Pareyson D, di Rocco M, Abicht A, McCourt E, Horvath R, Kölbel H, Larson A, Roos A, Yu TW, Finazzi D, Riganti C, Tolosano E, Chiabrando D.
https://pubmed.ncbi.nlm.nih.gov/41691085/

Novel NPRL3 variant associated with sleep-related hypermotor epilepsy: a case report and educational review.
Broggi S, Poppert KN, Mauritz M, Kalss G, Leitinger M, Abicht A, Trinka E, Rossini F.
https://pubmed.ncbi.nlm.nih.gov/41635699/

2025

Visual Diagnosis of Facioscapulohumeral Muscular Dystrophy (FSHD).
Erdmann H, Abicht A, Becker K.
https://pubmed.ncbi.nlm.nih.gov/40579967/

A 50-year cough: cerebellar ataxia, neuropathy, and vestibular areflexia syndrome (CANVAS).
Erdmann H, Abicht A, Strupp M.
Dtsch Arztebl Int 2025; 122: 167. https://doi.org/10.3238/arztebl.m2024.0247

Joint analysis of germline genetic data from over 29,000 cases with suspected hereditary breast and ovarian cancer (HBOC) as part of the NASGE initiative.
Henkel J, Laner A, Locher M, Wohlfrom T, Neitzel B, Becker K, Neuhann T, Abicht A, Steinke-Lange V, Klink B, Eichhorn B, Schmidt W, Berner D, Teubert A, Holtorf A, Heinrich S, Wildhardt G, Schulze M, von der Heyden L, Hörtnagel K, Steinberger D, Kleier S, Lorenz P, Glaubitz R, Biskup S, Holinski-Feder E.
https://pubmed.ncbi.nlm.nih.gov/39854808/

Umfassende Charakterisierung des D4Z4-Repeatarrays mittels Long-Read-Sequenzierung für eine präzise Diagnostik der Fazioskapulohumeralen Muskeldystrophie.
Scharf F, Erdmann H, Lucas MC, Gehling S, Benet-Pagès A, Schäfer J, Hallermayr A, Schönrock V, Köhler U, Neuhann T, Holinski-Feder E, Walter M, Schoser B, Abicht A.
Nervenheilkunde 2025; 44(03): 168-169. DOI: 10.1055/s-0044-1801528

Die Ränder des diagnostischen Spektrums der FSHD – Komplexe genetische Befunde in der Diagnostik der Fazioskapulohumeralen Muskeldystrophie (FSHD) und ihre Implikation für das molekulargenetische Modell der Erkrankung.
Erdmann H, Gehling S, Scharf F, Lucas MC, Kleefeld F, Becker K, Schönrock V, Saak A, Schäfer J, Neuhoff S, Hagenacker T, Porrmann J, Rausch HW, Schirmer L, Berking AC, Neuhann T, Holinski-Feder E, Walter M, Schoser B, Abicht A.
Nervenheilkunde 2025; 44(03): 143. DOI: 10.1055/s-0044-1801447

Blood biomarker fingerprints in a cohort of patients with CHRNE-related congenital myasthenic syndrome.
Della Marina A, Koutsoulidou A, Natera-de Benito D, Tykocinski LO, Tomazou M, Georgiou K, Laner A, Kölbel H, Nascimento A, Ortez C, Abicht A, Thakur BK, Lochmüller H, Phylactou LA, Ruck T, Schara-Schmidt U, Kale D, Hentschel A, Roos A.
https://pubmed.ncbi.nlm.nih.gov/39948634/

2024

Parallel in-depth analysis of repeat expansions: an updated Clin-CATS workflow for nanopore R10 flow cells.
Scholz V, Schönrock V, Erdmann H, Prokosch V, Schoedel M, Almus M, Sauer M, Mayer V, Breithausen E, van Buren I, Dineiger C, Heintz C, Hallermayr A, Neuhann T, Holinski-Feder E, Abicht A, Benet-Pagès A, Lucas MC.
bioRxiv 2024.11.05.622099; doi: https://doi.org/10.1101/2024.11.05.622099

Updated Structure of *CNBP* Repeat Expansions in Patients With Myotonic Dystrophy Type 2 and Its Implication for Standard Diagnostics.
Wendlandt M, Erdmann H, Rost S, Lucas MC, Becker K, Kleinle S, Timmer M, Bier A, Wunderlich G, Wenninger S, Walter MC, Neuhann T, Schoser B, Holinski-Feder E, Abicht A.
https://pubmed.ncbi.nlm.nih.gov/39703464/

A Novel MYH14 Variant Presenting as a New Phenotype of MYH14-Associated Neuromuscular Disorders-Clinicohistologic Findings and Review of the Literature.
Mensch A, Jordan B, Weis J, Nikolin S, Schneider I, Abicht A, Gehling S, Kendzierski T, Stoltenburg-Didinger G, Stoevesandt D, Kraya T, Zierz S, Naegel S.
https://pubmed.ncbi.nlm.nih.gov/39590923/

Analysis and occurrence of biallelic pathogenic repeat expansions in RFC1 in a German cohort of patients with a main clinical phenotype of motor neuron disease.
Schaub A, Erdmann H, Scholz V, Timmer M, Cordts I, Günther R, Reilich P, Abicht A, Schöberl F.
https://pubmed.ncbi.nlm.nih.gov/38916676/

LZTR1 loss-of-function variants associated with café au lait macules with or without freckling.
Horn S, Neuhann T, Hennig C, Abad-Perez A, Prott EC, Cardellini L, Potratz C, Leubner J, Eichhorn B, Merkel M, Abicht A, Kaindl AM.
https://pubmed.ncbi.nlm.nih.gov/39258154/

Optical Genome Mapping as a Potential Routine Clinical Diagnostic Method.
Barseghyan H, Eisenreich D, Lindt E, Wendlandt M, Scharf F, Benet-Pages A, Sendelbach K, Neuhann T, Abicht A, Holinski-Feder E, Koehler U.
https://pubmed.ncbi.nlm.nih.gov/38540401/

Novel Genetic and Biochemical Insights into the Spectrum of NEFL-Associated Phenotypes.
Della Marina A, Hentschel A, Czech A, Schara-Schmidt U, Preusse C, Laner A, Abicht A, Ruck T, Weis J, Choueiri C, Lochmüller H, Kölbel H, Roos A.
https://pubmed.ncbi.nlm.nih.gov/38578900/

A Homozygous NDUFS6 Variant Associated with Neuropathy and Optic Atrophy.
Gangfuß A, Rating P, Ferreira T, Hentschel A, Marina AD, Kölbel H, Sickmann A, Abicht A, Kraft F, Ruck T, Böhm J, Schänzer A, Schara-Schmidt U, Neuhann TM, Horvath R, Roos A.
https://pubmed.ncbi.nlm.nih.gov/38217609/

2023

Guideline for the management of myasthenic syndromes.
Wiendl H, Abicht A, Chan A, Della Marina A, Hagenacker T, Hekmat K, Hoffmann S, Hoffmann HS, Jander S, Keller C, Marx A, Melms A, Melzer N, Müller-Felber W, Pawlitzki M, Rückert JC, Schneider-Gold C, Schoser B, Schreiner B, Schroeter M, Schubert B, Sieb JP, Zimprich F, Meisel A.
https://pubmed.ncbi.nlm.nih.gov/38152089/

Novel Homozygous FA2H Variant Causing the Full Spectrum of Fatty Acid Hydroxylase-Associated Neurodegeneration (SPG35).
German A, Jukic J, Laner A, Arnold P, Socher E, Mennecke A, Schmidt MA, Winkler J, Abicht A, Regensburger M.
https://pubmed.ncbi.nlm.nih.gov/38275596/

Prospective evaluation of NGS-based sequencing in epilepsy patients: results of seven NASGE-associated diagnostic laboratories.
Witzel MGW, Gebhard C, Wenzel S, Kleier S, Eichhorn B, Lorenz P, von der Heyden L, Kuhn M, Luedeke M, Döcker M, Jüngling J, Schulte B, Hörtnagel K, Glaubitz R, Knippenberger S, Teubert A, Abicht A, Neuhann TM.
https://pubmed.ncbi.nlm.nih.gov/38125836/

Closing the Gap - Detection of 5q-Spinal Muscular Atrophy by Short-Read Next-Generation Sequencing and Unexpected Results in a Diagnostic Patient Cohort.
Kleinle S, Scholz V, Benet-Pagès A, Wohlfrom T, Gehling S, Scharf F, Rost S, Prott EC, Grinzinger S, Hotter A, Haug V, Niemeier S, Wiethoff-Ubrig L, Hagenacker T, Goldhahn K, von Moers A, Walter MC, Reilich P, Eggermann K, Kraft F, Kurth I, Erdmann H, Holinski-Feder E, Neuhann T, Abicht A.
https://pubmed.ncbi.nlm.nih.gov/37424474/

Reply: An epigenetic basis for genetic anticipation in facioscapulohumeral muscular dystrophy type 1.
Erdmann H, Scharf F, Hallermayr A, Barsegehyan H, Walter MC, Holinski-Feder E, Schoser B, Abicht A.
https://pubmed.ncbi.nlm.nih.gov/37348868/

Myofibrillar myopathy: a rare but important differential diagnosis of camptocormia in a patient with Parkinson's Disease.
Petry-Schmelzer JN, Abicht A, Barbe MT, Wunderlich G.
https://pubmed.ncbi.nlm.nih.gov/37287054/

Diagnostic yield and clinical relevance of expanded germline genetic testing for nearly 7000 suspected HBOC patients.
Henkel J, Laner A, Locher M, Wohlfrom T, Neitzel B, Becker K, Neuhann T, Abicht A, Steinke-Lange V, Holinski-Feder E.
https://pubmed.ncbi.nlm.nih.gov/37188824/

Parallel in-depth analysis of repeat expansions in ataxia patients by long-read sequencing
Hannes Erdmann, Florian Schöberl, Mădălina Giurgiu, Rafaela Magalhaes Leal Silva, Veronika Scholz, Florentine Scharf, Martin Wendlandt, Stephanie Kleinle, Marcus Deschauer, Georg Nübling, Wolfgang Heide, Sait Seymen Babacan, Christine Schneider, Teresa Neuhann, Katrin Hahn, Benedikt Schoser, Elke Holinski-Feder, Dieter A Wolf, Angela Abicht
https://pubmed.ncbi.nlm.nih.gov/36227727/

Methylation of the 4q35 D4Z4 repeat defines disease status in facioscapulohumeral muscular dystrophy
Hannes Erdmann, Florentine Scharf, Stefanie Gehling, Anna Benet-Pagès, Sibylle Jakubiczka, Kerstin Becker, Maria Seipelt, Felix Kleefeld, Karl Christian Knop, Eva-Christina Prott, Miriam Hiebeler, Federica Montagnese, Dieter Gläser, Matthias Vorgerd, Tim Hagenacker, Maggie C Walter, Peter Reilich, Teresa Neuhann, Martin Zenker, Elke Holinski-Feder, Benedikt Schoser, Angela Abicht
https://pubmed.ncbi.nlm.nih.gov/36100962/

The clinical and genetic spectrum of autosomal-recessive TOR1A-related disorders.
Saffari A, Lau T, Tajsharghi H, Karimiani EG, Kariminejad A, Efthymiou S, Zifarelli G, Sultan T, Toosi MB, Sedighzadeh S, Siu VM, Ortigoza-Escobar JD, AlShamsi AM, Ibrahim S, Al-Sannaa NA, Al-Hertani W, Sandra W, Tarnopolsky M, Alavi S, Li C, Day-Salvatore DL, Martínez-González MJ, Levandoski KM, Bedoukian E, Madan-Khetarpal S, Idleburg MJ, Menezes MJ, Siddharth A, Platzer K, Oppermann H, Smitka M, Collins F, Lek M, Shahrooei M, Ghavideldarestani M, Herman I, Rendu J, Faure J, Baker J, Bhambhani V, Calderwood L, Akhondian J, Imannezhad S, Mirzadeh HS, Hashemi N, Doosti M, Safi M, Ahangari N, Torbati PN, Abedini S, Salpietro V, Gulec EY, Eshaghian S, Ghazavi M, Pascher MT, Vogel M, Abicht A, Moutton S, Bruel AL, Rieubland C, Gallati S, Strom TM, Lochmüller H, Mohammadi MH, Alvi JR, Zackai EH, Keena BA, Skraban CM, Berger SI,

Andrew HE, Rahimian E, Morrow MM, Wentzensen IM, Millan F, Henderson LB, Dafsari HS, Jungbluth H, Gomez-Ospina N, McRae A, Peter M, Veltra D, Marinakis NM, Sofocleous C, Ashrafzadeh F, Pehlivan D, Lemke JR, Melki J, Benezit A, Bauer P, Weis D, Lupski JR, Senderek J, Christodoulou J, Chung WK, Goodchild R, Offiah AC, Moreno-De-Luca A, Mohnish S, Ebrahimi-Fakhari D, Houlden H, Maroofian R. The clinical and genetic spectrum of autosomal-recessive TOR1A-related disorders.
https://pubmed.ncbi.nlm.nih.gov/36757831/

2022

Persistent hypokalaemia and intermittent muscle weakness.
Rabenstein M, Abicht A, Brunn A, Lehmann H, Wunderlich G.
https://pubmed.ncbi.nlm.nih.gov/35907633/

Parallel in-depth analysis of repeat expansions in ataxia patients by long-read sequencing.
Erdmann H, Schöberl F, Giurgiu M, Leal Silva RM, Scholz V, Scharf F, Wendlandt M, Kleinle S, Deschauer M, Nübling G, Heide W, Babacan SS, Schneider C, Neuhann T, Hahn K, Schoser B, Holinski-Feder E, Wolf DA, Abicht A.
https://pubmed.ncbi.nlm.nih.gov/36227727/

Methylation of the 4q35 D4Z4 repeat defines disease status in facioscapulohumeral muscular dystrophy
Hannes Erdmann, Florentine Scharf, Stefanie Gehling, Anna Benet-Pagès, Sibylle Jakubiczka, Kerstin Becker, Maria Seipelt , Felix Kleefeld , Karl Christian Knop, Eva Christina Prott, Miriam Hiebeler, Federica Montagnese, Dieter Gläser, Matthias Vorgerd , Tim Hagenacker, Maggie C Walter, Peter Reilich, Teresa Neuhan, Martin Zenker, Elke Holinski-Feder, Benedikt Schoser, Angela Abicht
https://pubmed.ncbi.nlm.nih.gov/36100962/

Persistent hypokalaemia and intermittent muscle weakness
Monika Rabenstein, Angela Abicht, Anna Brunn, Helmar Lehmann, Gilbert Wunderlich
https://pubmed.ncbi.nlm.nih.gov/35907633/

O'Donnell-Luria-Rodan syndrome: description of a second multinational cohort and refinement of the phenotypic spectrum
Clara Velmans, Anne H O'Donnell-Luria , Emanuela Argilli, Frederic Tran Mau-Them, Antonio Vitobello , Marcus Cy Chan, Jasmine Lee-Fong Fung, Megan Rech, Angela Abicht, Marion Aubert Mucca, Jason Carmichael, Nicolas Chassaing, Robin Clark, Christine Coubes, Anne-Sophie Denommé-Pichon, John Karl de Dios, Eleina England, Benoit Funalot, Marion Gerard, Maries Joseph, Colleen Kennedy, Camille Kumps, Marjolaine Willems, Ingrid M B H van de Laar, Coranne Aarts-Tesselaar, Marjon van Slegtenhorst, Daphné Lehalle, Kathleen Leppig, Lennart Lessmeier, Lynn S Pais, Heather Paterson, Subhadra Ramanathan, Lance H Rodan, Andrea Superti-Furga, Brian H Y Chung, Elliott Sherr, Christian Netzer, Christian P Schaaf, Florian Erger
https://pubmed.ncbi.nlm.nih.gov/34321323/

Identification of a novel homozygous synthesis of cytochrome c oxidase 2 variant in siblings with early-onset axonal Charcot-Marie-Tooth disease
Andrea Gangfuß, Andreas Hentschel, Nina Rademacher, Albert Sickmann, Burkhard Stüve, Rita Horvath, Claudia Gross, Nicolai Kohlschmidt, Fabian Förster, Angela Abicht, Anne Schänzer, Ulrike Schara-Schmidt, Andreas Roos, Adela Della Marina
https://pubmed.ncbi.nlm.nih.gov/35112411/

Sensory neuropathy due to RFC1 in a patient with ALS: more than a coincidence?
Florian Schoeberl, Angela Abicht, Clemens Kuepper , Stefanie Voelk , Stefan Sonnenfeld , Matthias Tonon , Annalisa Schaub, Veronika Scholz , Stephanie Kleinle , Hannes Erdmann , Dieter A Wolf , Peter Reilich
https://pubmed.ncbi.nlm.nih.gov/34821988/

Congenital myopathy and epidermolysis bullosa due to PLEC variant
Maggie C Walter, Peter Reilich, Sabine Krause, Miriam Hiebeler, Stefanie Gehling, Hans H Goebel, Benedikt Schoser, Angela Abicht
https://doi.org/10.1016/j.nmd.2021.09.009

Cutaneous T-cell lymphoma mimicking myopathy with lipoatrophy
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https://doi.org/10.1016/j.nmd.2021.11.009

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Gross, Nicolai Kohlschmidt, Fabian Förster, Angela Abicht, Anne Schänzer, Ulrike Schara-Schmidt, Andreas Roos, Adela Della Marina
https://doi.org/10.1002/humu.24338

Initial Clinical Experience with NIPT for Rare Autosomal Aneuploidies and Large Copy Number Variations
Thomas Harasim, Teresa Neuhann, Anne Behnecke, Miriam Stampfer, Elke Holinski-Feder, Angela Abicht
https://doi.org/10.3390/jcm11020372

2021

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https://doi.org/10.1016/j.nmd.2021.11.009

Effect of Discontinuation of Nusinersen Treatment in Long-Standing SMA_3
Miriam Hiebeler, Angela Abicht, Peter Reilich, Maggie C Walter
https://doi.org/10.3233/jnd-210644

Sensory neuropathy due to RFC_1 in a patient with ALS: more than a coincidence?
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https://doi.org/10.1007/s00415-021-10835-9

Location matters – Genotype – phenotype correlation in $LRSAM_1$ mutations associated with rare Charcot-Marie-Tooth neuropathy CMT_2P
Peter Reilich, Beate Schlotter, Federica Montagnese, Berit Jordan, Friedrich Stock, Mario Schäff-Vogelsang, Benjamin Hotter, Katherina Eger, Isabel Diebold, Hannes Erdmann, Kerstin Becker, Ulrike Schön, Angela Abicht
https://doi.org/10.1016/j.nmd.2020.11.011

Next Generation Sequencing in Pediatric Epilepsy Using Customized Panels: Size Matters
Eva-Katharina Willimsky, Anna Munzig, Karin Mayer, Saskia Biskup, Angela Abicht, Konstanze Hoertnagel, Hubertus von Voss, Hanns-Georg Klein, Imma Rost, Line H G Larsen, Hanns Atli Dahl, Hannes Hoelz, Celina von Stuelpnagel, Ingo Borggraefe
https://doi.org/10.1055/s-0040-1712488

Thiamine Treatment and Favorable Outcome in an Infant with Biallelic TPK_1 Variants
Matthias Eckenweiler, Johannes A Mayr, Sarah Grünert, Angela Abicht, Rudolf Korinthenberg
https://doi.org/10.1055/s-0040-1715631

Congenital myopathy and epidermolysis bullosa due to PLEC variant
Maggie C Walter, Peter Reilich, Sabine Krause, Miriam Hiebeler, Stefanie Gehling, Hans H Goebel, Benedikt Schoser, Angela Abicht
https://doi.org/10.1016/j.nmd.2021.09.009

Differential diagnosis of vacuolar myopathies in the NGS era.
Mair D, Biskup S, Kress W, Abicht A, Brück W, Zechel S, Knop KC, Koenig FB, Tey S, Nikolin S, Eggermann K, Kurth I, Ferbert A, Weis J. Brain Pathol. 2020 Sep;30(5):877-896.
https://doi.org/10.1111/bpa.12864

Rare intronic mutation between Exon 62 and 63 (c.9225-285A>G) of the dystrophin gene associated with atypical BMD phenotype.
Schüssler SC, Gerhalter T, Abicht A, Müller-Felber W, Nagel AM, Trollmann R., Neuromuscul Disord. 2020 Aug;30(8):680-684.
https://doi.org/10.1016/j.nmd.2020.06.003

O'Donnell-Luria-Rodan syndrome: description of a second multinational cohort and refinement of the phenotypic spectrum
Clara Velmans, Anne H O'Donnell-Luria, Emanuela Argilli, Frederic Tran Mau-Them, Antonio Vitobello, Marcus Cy Chan, Jasmine Lee-Fong Fung, Megan Rech, **Angela Abicht**, Marion Aubert Mucca, Jason Carmichael, Nicolas Chassaing, Robin Clark, Christine Coubes, Anne-Sophie Denommé-Pichon, John Karl de Dios, Eleina England, Benoit Funalot, Marion Gerard, Maries Joseph, Colleen Kennedy, Camille Kumps, Marjolaine Willems, Ingrid M B H van de Laar, Coranne Aarts-Tesselaar, Marjon van Slegtenhorst, Daphné Lehalle, Kathleen

Leppig, Lennart Lessmeier, Lynn S Pais, Heather Paterson, Subhadra Ramanathan, Lance H Rodan, Andrea Superti-Furga, Brian H Y Chung, Elliott Sherr, Christian Netzer, Christian P Schaaf, Florian Erger. https://doi.org/10.1136/jmedgenet-2020-107470

Location matters - Genotype-phenotype correlation in LRSAM1 mutations associated with rare Charcot-Marie-Tooth neuropathy CMT2P.
P. Reilich, B. Schlotter, F. Montagnese, B. Jordan, F. Stock, M. Schäff-Vogelsang, B. Hotter, K. Eger, I. Diebold, H. Erdmann, K. Becker, U. Schön, & A. Abicht, Neuromuscular disorders: NMD, 31 (2021) 123–133. https://doi.org/10.1016/j.nmd.2020.11.011.

Actionable secondary findings in arrhythmogenic right ventricle cardiomyopathy genes: impact and challenge of genetic counseling.
A. Abicht, U. Schön, A. Laner, E. Holinski-Feder, & I. Diebold, Cardiovascular Diagnosis and Therapy, 11 (2021) 637–649. https://doi.org/10.21037/cdt-20-585

Effect of Discontinuation of Nusinersen Treatment in Long-Standing SMA3.
M. Hiebeler, A. Abicht, P. Reilich, & M. C. Walter, Journal of Neuromuscular Diseases, (2021). https://content.iospress.com/articles/journal-of-neuromuscular-diseases/jnd210644

2020

Long Term Follow-Up on Pediatric Cases With Congenital Myasthenic Syndromes-A Retrospective Single Centre Cohort Study. Della Marina A, Wibbeler E, Abicht A, Kölbel H, Lochmüller H, Roos A, Schara U. *Front Hum Neurosci.* 2020 Dec 7;14:560860. https://doi.org/10.3389/fnhum.2020.560860

Thiamine Treatment and Favorable Outcome in an Infant with Biallelic TPK1 Variants. Eckenweiler M, Mayr JA, Grünert S, Abicht A, Korinthenberg R. *Neuropediatrics.* 2020 Oct 21. https://doi.org/10.1055/s-0040-1715631

Next Generation Sequencing in Pediatric Epilepsy Using Customized Panels: Size Matters. Willimsky EK, Munzig A, Mayer K, Biskup S, Abicht A, Hoertnagel K, Voss HV, Klein HG, Rost I, Larsen LHG, Dahl HA, Hoelz H, Stuelpnagel CV, Borggraefe I. *Neuropediatrics*. 2020 Oct 21. https://doi.org/10.1055/s-0040-1712488

Differential diagnosis of vacuolar myopathies in the NGS era. Mair D, Biskup S, Kress W, Abicht A, Brück W, Zechel S, Knop KC, Koenig FB, Tey S, Nikolin S, Eggermann K, Kurth I, Ferbert A, Weis J. *Brain Pathol*. 2020;30(5):877-896. https://doi.org/10.1111/bpa.12864

Dilative cardiomyopathy displaying double trouble etiology: Myocarditis and Mcleod syndrome? Montagnese F, Grabmaier U, Abicht A, Schoser B. *Clin Neurol Neurosurg*. 2020;197:106122. https://doi.org/10.1016/j.clineuro.2020.106122

Rare intronic mutation between Exon 62 and 63 (c.9225-285A>G) of the dystrophin gene associated with atypical BMD phenotype. Schüssler SC, Gerhalter T, Abicht A, Müller-Felber W, Nagel AM, Trollmann R. *Neuromuscul Disord*. 2020;30(8):680-684. https://doi.org/10.1016/j.nmd.2020.06.003

Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. Boonsimma P, Michael Gasser M, Netbaramee W, Wechapinan T, Srichomthong C, Ittiwut C, Wagner M, Krenn M, Zimprich F, Abicht A, Biskup S, Roser T, Borggraefe I, Suphapeetiporn K, Shotelersuk V. *Gene*. 2020;749:144709. https://doi.org/10.1016/j.gene.2020.144709

Genotype-phenotype correlation in a novel ABHD12 mutation underlying PHARC syndrome. Thimm A, Rahal A, Schoen U, Abicht A, Klebe S, Kleinschnitz C, Hagenacker T, Stettner M. *J Peripher Nerv Syst*. 2020;25(2):112-116. https://doi.org/10.1111/jns.12367

Critical assessment of secondary findings in genes linked to primary arrhythmia syndromes. Diebold I, Schön U, Scharf F, Benet-Pagès A, Laner A, Holinski-Feder E, Abicht A. *Hum Mutat*. 2020;41(5):1025-1032. https://doi.org/10.1002/humu.23996

[Multiple acyl-CoA dehydrogenase deficiency/glutaric aciduria type 2: difficult diagnosis, easy to treat]. Rabenstein M, Weis J, Abicht A, Fink GR, Lehmann HC, Wunderlich G. *Nervenarzt*. 2020;91(4):349-352. https://doi.org/10.1007/s00115-020-00886-0

Delineating MT-ATP6-associated disease: From isolated neuropathy to early onset neurodegeneration. Stendel C, Neuhofer C, Floride E, Yuqing S, Ganetzky RD, Park J, Freisinger P, Kornblum C, Kleinle S, Schöls L, Distelmaier F, Stettner GM, Büchner B, Falk MJ, Mayr JA, Synofzik M, Abicht A, Haack TB, Prokisch H, Wortmann SB, Murayama K, Fang F, Klopstock T. *Neurol Genet*. 2020;6(1):e393. https://doi.org/10.1212/NXG.0000000000000393

Cooccurrence of Two Different Genetic Diseases: A Case of Valproic Acid Hepatotoxicity in Nicolaides-Baraitser Syndrome (SMARCA2 Mutation)-Due to a POLG1-Related Effect? Hofmeister B, von Stülpnagel C, Berweck S, Abicht A, Kluger G, Weber P. *Neuropediatrics*. 2020;51(1):49-52. https://doi.org/10.1055/s-0039-1694976

ATP1A3-related epilepsy: Report of seven cases and literature-based analysis of treatment response. Gasser M, Boonsimma P, Netbaramee W, Wechapinan T, Srichomthomg C, Ittiwut C, Krenn M, Zimprich F, Milenkovic I, Abicht A, Biskup S, Roser T, Shotelersuk V, Tacke M, Kuersten M, Wagner M, Borggraefe I, Suphapeetiporn K, von Stülpnagel C. *J Clin Neurosci*. 2020;72:31-38. https://doi.org/10.1016/j.jocn.2020.01.041

Impact on Clinical Decision Making of Next-Generation Sequencing in Pediatric Epilepsy in a Tertiary Epilepsy Referral Center. Hoelz H, Herdl C, Gerstl L, Tacke M, Vill K, von Stuelpnagel C, Rost I, Hoertnagel K, Abicht A, Hollizeck S, Larsen LHG, Borggraefe I. *Clin EEG Neurosci*. 2020;51(1):61-69. https://doi.org/10.1177/1550059419876518

2019

VLDLR-associated Pontocerebellar Hypoplasia with Nonprogressive Congenital Ataxia and a Diagnostic Neuroimaging Pattern. Wilker M, Christen H-J, Schuster S, Abicht A, Boltshauser E. *Neuropediatrics*. 2019;50(6):404-405. https://doi.org/10.1055/s-0039-1688953

Charcot-Marie-Tooth disease type 2CC due to a frameshift mutation of the neurofilament heavy polypeptide gene in an Austrian family. Ikenberg E, Reilich P, **Abicht A**, Heller C, Schoser B, Walter MC. *Neuromuscul Disord*. 2019;29(5):392-397. https://doi.org/10.1016/j.nmd.2019.02.007

Congenital myasthenic syndrome caused by novel COL13A1 mutations. Dusl M, Moreno T, Munell F, Macaya A, Gratacòs M, Abicht A, Strom TM, Lochmüller H, Senderek J. *J Neurol*. 2019;266(5):1107-1112. https://doi.org/10.1007/s00415-019-09239-7

HADHA and HADHB gene associated phenotypes - Identification of rare variants in a patient cohort by Next Generation Sequencing. Diebold I, Schön U, Horvath R, Schwartz O, Holinski-Feder E, Kölbel H, **Abicht A**. *Mol Cell Probes*. 2019;44:14-20. https://doi.org/10.1016/j.mcp.2019.01.003

Characteristic clinical and ultrastructural findings in nesprinopathies. Kölbel H, Abicht A, Schwartz O, Katona I, Paulus W, Neuen-Jacob E, Weis J, Schara U. *Eur J Paediatr Neurol*. 2019;23(2):254-261. https://doi.org/10.1016/j.ejpn.2018.12.011

Congenital myasthenic syndromes in adulthood : Challenging, rare but treatable. Wunderlich G, Abicht A, Brunn A, Daimagüler H-S, Schroeter M, Fink GR, Lehmann HC, Cirak S. *Nervenarzt*. 2019;90(2):148-159. https://doi.org/10.1007/s00115-018-0562-9

A Novel Gain-of-Function Nav1.9 Mutation in a Child With Episodic Pain. Huang J, Estacion M, Zhao P, Dib-Hajj FB, Schulman B, Abicht A, Kurth I, Brockmann K, Waxman SG, Dib-Hajj SD. *Front Neurosci*. 2019;13:918. https://doi.org/10.3389/fnins.2019.00918

2018

A nomenclature and classification for the congenital myasthenic syndromes: preparing for FAIR data in the genomic era. Thompson R, Abicht A, Beeson D, Engel AG, Eymard B, Maxime E, Lochmüller H. *Orphanet J Rare Dis*. 2018;13(1):211. https://doi.org/10.1186/s13023-018-0955-7

Mitochondrial and nuclear disease panel (Mito-aND-Panel): Combined sequencing of mitochondrial and nuclear DNA by a cost-effective and sensitive NGS-based method. Abicht A, Scharf F, Kleinle S, Schön U, Holinski-Feder E, Horvath R, Benet-Pagès A, Diebold I. *Mol Genet Genomic Med*. 2018;6(6):1188-1198. https://doi.org/10.1002/mgg3.500

The Curse of Apneic Spells. Radke J, Dreesmann M, Radke M, von Moers A, Abicht A, Stenzel W, Goebel HH. *Semin Pediatr Neurol*. 2018;26:56-58. https://doi.org/10.1016/j.spen.2017.03.006

A new case of limb girdle muscular dystrophy 2G in a Greek patient, founder effect and review of the literature. Brusa R, Magri F, Papadimitriou D, Govoni A, Del Bo R, Ciscato P, Savarese M, Cinnante C, Walter MC, Abicht A, Bulst S, Corti S, Moggio M, Bresolin N, Nigro V, Comi GP. *Neuromuscul Disord*. 2018;28(6):532-537. https://doi.org/10.1016/j.nmd.2018.04.006

Extension of the phenotype of biallelic loss-of-function mutations in SLC25A46 to the severe form of pontocerebellar hypoplasia type I. Braunisch MC, Gallwitz H, Abicht A, Diebold I, Holinski-Feder E, Van

Maldergem L, Lammens M, Kovács-Nagy R, Alhaddad B, Strom TM, Meitinger T, Senderek J, Rudnik-Schöneborn S, Haack TB. *Clin Genet*. 2018;93(2):255-265. https://doi.org/10.1111/cge.13084

Congenital myasthenic syndrome with episodic apnoea: clinical, neurophysiological and genetic features in the long-term follow-up of 19 patients. McMacken G, Whittaker RG, Evangelista T, Abicht A, Dusl M, Lochmüller H. *J Neurol*. 2018;265(1):194-203. https://doi.org/10.1007/s00415-017-8689-3

2017

Molecular characterization of congenital myasthenic syndromes in Spain. Natera-de Benito D, Töpf A, Vilchez JJ, González-Quereda L, Domínguez-Carral J, Díaz-Manera J, Ortez C, Bestué M, Gallano P, Dusl M, Abicht A, Müller JS, Senderek J, García-Ribes A, Muelas N, Evangelista T, Azuma Y, McMacken G, Paipa Merchan A, Rodríguez Cruz PM, Camacho A, Jiménez E, Miranda-Herrero MC, Santana-Artiles A, García-Campos O, Dominguez-Rubio R, Olivé M, Colomer J, Beeson D, Lochmüller H, Nascimento A. *Neuromuscul Disord*. 2017;27(12):1087-1098. https://doi.org/10.1016/j.nmd.2017.08.003

Corrigendum to "Rare diagnosis of telethoninopathy (LGMD2G) in a Turkish patient" [Neuromuscular Disorders 27 (2017) 856-860]. Ikenberg E, Karin I, Ertl-Wagner B, Abicht A, Bulst S, Krause S, Schoser B, Reilich P, Walter MC. *Neuromuscul Disord*. 2017;27(12):e1. https://doi.org/10.1016/j.nmd.2017.10.001

Rare diagnosis of telethoninopathy (LGMD2G) in a Turkish patient. Ikenberg E, Karin I, Ertl-Wagner B, Abicht A, Bulst S, Krause S, Schoser B, Reilich P, Walter MC. *Neuromuscul Disord*. 2017;27(9):856-860. https://doi.org/10.1016/j.nmd.2017.05.017

The Increasing Genetic and Phenotypical Diversity of Congenital Myasthenic Syndromes. McMacken G, Abicht A, Evangelista T, Spendiff S, Lochmüller H. *Neuropediatrics*. 2017;48(4):294-308. https://doi.org/10.1055/s-0037-1602832

Lifetime exercise intolerance with lactic acidosis as key manifestation of novel compound heterozygous ACAD9 mutations causing complex I deficiency. Schrank B, Schoser B, Klopstock T, Schneiderat P, Horvath R, Abicht A, Holinski-Feder E, Augustis S. *Neuromuscul Disord*. 2017;27(5):473-476. https://doi.org/10.1016/j.nmd.2017.02.005

Stroke as Initial Manifestation of Adenosine Deaminase 2 Deficiency. Elbracht M, Mull M, Wagner N, Kuhl C, Abicht A, Kurth I, Tenbrock K, Häusler M. *Neuropediatrics*. 2017;48(2):111-114. https://doi.org/10.1055/s-0036-1597611

2016

Novel homozygous RARS2 mutation in two siblings without pontocerebellar hypoplasia - further expansion of the phenotypic spectrum. Lühl S, Bode H, Schlötzer W, Bartsakoulia M, Horvath R, Abicht A, Stenzel M, Kirschner J, Grünert SC. *Orphanet J Rare Dis*. 2016;11(1):140. https://doi.org/10.1186/s13023-016-0525-9

Identification of mutations in the MYO9A gene in patients with congenital myasthenic syndrome. O'Connor E, Töpf A, Müller JS, Cox D, Evangelista T, Colomer J, Abicht A, Senderek J, Hasselmann O, Yaramis A, Laval SH, Lochmüller H. *Brain*. 2016;139(Pt 8):2143-2153. https://doi.org/10.1093/brain/aww130

Novel genetic and neuropathological insights in neurogenic muscle weakness, ataxia, and retinitis pigmentosa (NARP). Claeys KG, Abicht A, Häusler M, Kleinle S, Wiesmann M, Schulz JB, Horvath R, Weis J. *Muscle Nerve*. 2016;54(2):328-333. https://doi.org/10.1002/mus.25125

Mitochondrial dysfunction in liver failure requiring transplantation. Lane M, Boczonadi V, Bachtari S, Gomez-Duran A, Langer T, Griffiths A, Kleinle S, Dineiger C, Abicht A, Holinski-Feder E, Schara U, Gerner P, Horvath R. *J Inherit Metab Dis*. 2016;39(3):427-436. https://doi.org/10.1007/s10545-016-9927-z

Thomsen myotonia--A 4-generation family with a new mutation and a mild phenotype. Derevenciuc A-I, Abicht A, Hamza S, Roth C, Ferbert A. *Muscle Nerve*. 2016;53(4):653-654. https://doi.org/10.1002/mus.24971

The Variant p.(Arg183Trp) in SPTLC2 Causes Late-Onset Hereditary Sensory Neuropathy. Suriyanarayanan S, Auranen M, Toppila J, Paetau A, Shcherbii M, Palin E, Wei Y, Lohioja T, Schlotter-Weigel B, Schön U, **Abicht A**, Rautenstrauss B, Tyynismaa H, Walter MC, Hornemann T, Ylikallio E. *Neuromolecular Med*. 2016;18(1):81-90. https://doi.org/10.1007/s12017-015-8379-1

KLHL40-related nemaline myopathy with a sustained, positive response to treatment with acetylcholinesterase inhibitors. Natera-de Benito D, Nascimento A, **Abicht A**, Ortez C, Jou C, Müller JS, Evangelista T, Töpf A, Thompson R, Jimenez-Mallebrera C, Colomer J, Lochmüller H. *J Neurol*. 2016;263(3):517-523. https://doi.org/10.1007/s00415-015-8015-x

2015

Long-term follow-up in patients with congenital myasthenic syndrome due to RAPSN mutations. Natera-de Benito D, Bestué M, Vilchez JJ, Evangelista T, Töpf A, García-Ribes A, Trujillo-Tiebas MJ, García-Hoyos M, Ortez C, Camacho A, Jiménez E, Dusl M, **Abicht A**, Lochmüller H, Colomer J, Nascimento A. *Neuromuscul Disord*. 2016;26(2):153-159. https://doi.org/10.1016/j.nmd.2015.10.013

Early-onset leukoencephalopathy due to a homozygous missense mutation in the DARS2 gene. Köhler C, Heyer C, Hoffjan S, Stemmler S, Lücke T, Thiels C, Kohlschütter A, Löbel U, Horvath R, Kleinle S, Benet-Pages A, **Abicht A**. *Mol Cell Probes*. 2015;29(5):319-322. https://doi.org/10.1016/j.mcp.2015.06.005

A 3'-UTR mutation creates a microRNA target site in the GFPT1 gene of patients with congenital myasthenic syndrome. Dusl M, Senderek J, Müller JS, Vogel JG, Pertl A, Stucka R, Lochmüller H, David R, **Abicht A**. *Hum Mol Genet*. 2015;24(12):3418-3426. https://doi.org/10.1093/hmg/ddv090

A de novo Mutation in the SCN4A Gene Causing Sodium Channel Myotonia. Ørstavik K, Wallace SC, Torbergsen T, **Abicht A**, Erik Tangsrud S, Kerty E, Rasmussen M. *J Neuromuscul Dis*. 2015;2(2):181-184. https://doi.org/10.3233/JND-150069

Respiratory chain deficiency in nonmitochondrial disease. Pyle A, Nightingale HJ, Griffin H, **Abicht A**, Kirschner J, Baric I, Cuk M, Douroudis K, Feder L, Kratz M, Czermin B, Kleinle S, Santibanez-Koref M, Karcagi V, Holinski-Feder E, Chinnery PF, Horvath R. *Neurol Genet*. 2015;1(1):e6. https://doi.org/10.1212/NXG.0000000000000006

2014

ATP synthase deficiency due to TMEM70 mutation leads to ultrastructural mitochondrial degeneration and is amenable to treatment. Braczynski AK, Vlaho S, Müller K, Wittig I, Blank A-E, Tews DS, Drott U, Kleinle S, **Abicht A**, Horvath R, Plate KH, Stenzel W, Goebel HH, Schulze A, Harter PN, Kieslich M, Mittelbronn M. *Biomed Res Int*. 2015;2015:462592. https://doi.org/10.1155/2015/462592

Agrin mutations lead to a congenital myasthenic syndrome with distal muscle weakness and atrophy. Nicole S, Chaouch A, Torbergsen T, Bauché S, de Bruyckere E, Fontenille M-J, Horn MA, van Ghelue M, Løseth S, Issop Y, Cox D, Müller JS, Evangelista T, Stålberg E, Ioos C, Barois A, Brochier G, Sternberg D, Fournier E, Hantaï D, **Abicht A**, Dusl M, Laval SH, Griffin H, Eymard B, Lochmüller H. *Brain*. 2014;137(Pt 9):2429-2443. https://doi.org/10.1093/brain/awu160

Use of whole-exome sequencing to determine the genetic basis of multiple mitochondrial respiratory chain complex deficiencies. 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 AAM, Vassallo G, Gorman GS, Ramesh V, Turnbull DM, Santibanez-Koref M, McFarland R, Horvath R, Chinnery PF. *JAMA*. 2014;312(1):68-77. https://doi.org/10.1001/jama.2014.7184

Novel ETFDH mutation and imaging findings in an adult with glutaric aciduria type II. Rosenbohm A, Süssmuth SD, Kassubek J, Müller H-P, Pontes C, **Abicht A**, Bulst S, Ludolph AC, Pinkhardt E. *Muscle Nerve*. 2014;49(3):446-450. https://doi.org/10.1002/mus.23979

Congenital myasthenic syndrome due to choline acetyltransferase mutations in infants: clinical suspicion and comprehensive electrophysiological assessment are important for early diagnosis. Dilena R, **Abicht A**, Sergi P, Comi GP, Di Fonzo A, Chidini G, Natacci F, Barbieri S, Lochmüller H. *J Child Neurol*. 2014;29(3):389-393. https://doi.org/10.1177/0883073812470000

Novel TPM3 mutation in a family with cap myopathy and review of the literature. Schreckenbach T, Schröder JM, Voit T, **Abicht A**, Neuen-Jacob E, Roos A, Bulst S, Kuhl C, Schulz JB, Weis J, Claeys KG. *Neuromuscul Disord*. 2014;24(2):117-124. https://doi.org/10.1016/j.nmd.2013.10.002

Salbutamol-responsive limb-girdle congenital myasthenic syndrome due to a novel missense mutation and heteroallelic deletion in MUSK. Gallenmüller C, Müller-Felber W, Dusl M, Stucka R, Guergueltcheva V, Blaschek A, von der Hagen M, Huebner A, Müller JS, Lochmüller H, **Abicht A**. *Neuromuscul Disord*. 2014;24(1):31-35. https://doi.org/10.1016/j.nmd.2013.08.002

Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Chaouch A, Porcelli V, Cox D, Edvardson S, Scarcia P, De Grassi A, Pierri CL, Cossins J, Laval SH, Griffin H, Müller JS, Evangelista T, Töpf A, **Abicht A**, Huebner A, von der Hagen M, Bushby K, Straub V, Horvath R, Elpeleg O, Palace J, Senderek J, Beeson D, Palmieri L, Lochmüller H. *J Neuromuscul Dis*. 2014;1(1):75-90. https://doi.org/10.3233/JND-140021

2013

Novel CACNA1A mutation(s) associated with slow saccade velocities. Kipfer S, Jung S, Lemke JR, Kipfer-Kauer A, Howell JP, Kaelin-Lang A, Nyffeler T, Gutbrod K, **Abicht A**, Müri RM. *J Neurol*. 2013;260(12):3010-3014. https://doi.org/10.1007/s00415-013-7099-4

Quinine sulfate as a therapeutic option in a patient with slow channel congenital myasthenic syndrome. Peyer A-K, **Abicht A**, Heinimann K, Sinnreich M, Fischer D. *Neuromuscul Disord*. 2013;23(7):571-574. https://doi.org/10.1016/j.nmd.2013.04.001

DOK7 limb-girdle myasthenic syndrome mimicking congenital muscular dystrophy. Mahjneh I, Lochmüller H, Muntoni F, **Abicht A**. *Neuromuscul Disord*. 2013;23(1):36-42. https://doi.org/10.1016/j.nmd.2012.06.355

NDUFS8-related Complex I Deficiency Extends Phenotype from "PEO Plus" to Leigh Syndrome. Marina AD, Schara U, Pyle A, Möller-Hartmann C, Holinski-Feder E, **Abicht A**, Czermin B, Lochmüller H, Griffin H, Santibanez-Koref M, Chinnery PF, Horvath R. *JIMD Rep*. 2013;10:17-22. https://doi.org/10.1007/8904_2012_195

2012

Exercise-induced myalgia and rhabdomyolysis in a patient with the rare m.3243A>T mtDNA mutation. Czell D, **Abicht A**, Hench J, Weber M. *BMJ Case Rep*. 2012;2012. https://doi.org/10.1136/bcr-2012-006980

What is influencing the phenotype of the common homozygous polymerase-γ mutation p.Ala467Thr? Neeve VCM, Samuels DC, Bindoff LA, van den Bosch B, Van Goethem G, Smeets H, Lombès A, Jardel C, Hirano M, Dimauro S, De Vries M, Smeitink J, Smits BW, de Coo IFM, Saft C, Klopstock T, Keiling B-C, Czermin B, **Abicht A**, Lochmüller H, Hudson G, Gorman GG, Turnbull DM, Taylor RW, Holinski-Feder E, Chinnery PF, Horvath R. *Brain*. 2012;135(Pt 12):3614-3626. https://doi.org/10.1093/brain/aws298

Congenital myasthenic syndromes: achievements and limitations of phenotype-guided gene-after-gene sequencing in diagnostic practice: a study of 680 patients. Abicht A, Dusl M, Gallenmüller C, Guergueltcheva V, Schara U, Della Marina A, Wibbeler E, Almaras S, Mihaylova V, von der Hagen M, Huebner A, Chaouch A, Müller JS, Lochmüller H. *Hum Mutat*. 2012;33(10):1474-1484. https://doi.org/10.1002/humu.22130

In vitro supplementation with deoxynucleoside monophosphates rescues mitochondrial DNA depletion. Bulst S, Holinski-Feder E, Payne B, **Abicht A**, Krause S, Lochmüller H, Chinnery PF, Walter MC, Horvath R. *Mol Genet Metab*. 2012;107(1-2):95-103. https://doi.org/10.1016/j.ymgme.2012.04.022

Febrile infection-related epilepsy syndrome without detectable autoantibodies and response to immunotherapy: a case series and discussion of epileptogenesis in FIRES. van Baalen A, Häusler M, Plecko-Startinig B, Strautmanis J, Vlaho S, Gebhardt B, Rohr A, **Abicht A**, Kluger G, Stephani U, Probst C, Vincent A, Bien CG. *Neuropediatrics*. 2012;43(4):209-216. https://doi.org/10.1055/s-0032-1323848

Congenital myasthenic syndromes: current diagnostic and therapeutic approaches. Schara U, Della Marina A, **Abicht A**. *Neuropediatrics*. 2012;43(4):184-193. https://doi.org/10.1055/s-0032-1323850

Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. 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. *J Neurol*. 2012;259(5):838-850. https://doi.org/10.1007/s00415-011-6262-z

Classical MERRF phenotype associated with mitochondrial tRNA(Leu) (m.3243A>G) mutation. Brackmann F, **Abicht A**, Ahting U, Schröder R, Trollmann R. *Eur J Pediatr*. 2012;171(5):859-862. https://doi.org/10.1007/s00431-011-1662-8

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*. 2012;259(3):474-481. https://doi.org/10.1007/s00415-011-6204-9

2011

Neuromuscular signal transmission in adulthood. Current facets of acquired and hereditary disorders. Abicht A, Kröger S, Schoser B. *Nervenarzt*. 2011;82(6):707-711. https://doi.org/10.1007/s00115-010-2969-9

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;88(2):162-172. https://doi.org/10.1016/j.ajhg.2011.01.008

Acute liver failure with subsequent cirrhosis as the primary manifestation of TRMU mutations. Schara U, von Kleist-Retzow J-C, Lainka E, Gerner P, Pyle A, Smith PM, Lochmüller H, Czermin B, **Abicht A**, Holinski-Feder E, Horvath R. *J Inherit Metab Dis*. 2011;34(1):197-201. https://doi.org/10.1007/s10545-010-9250-z

Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. Kemp JP, Smith PM, Pyle A, Neeve VCM, Tuppen HAL, Schara U, Talim B, Topaloglu H, Holinski-Feder E, **Abicht A**, Czermin B, Lochmüller H, McFarland R, Chinnery PF, Chrzanowska-Lightowlers ZMA, Lightowlers RN, Taylor RW, Horvath R. *Brain*. 2011;134(Pt 1):183-195. https://doi.org/10.1093/brain/awq320

2010

Clinical and neuropathological findings in patients with TACO1 mutations. Seeger J, Schrank B, Pyle A, Stucka R, Lörcher U, Müller-Ziermann S, **Abicht A**, Czermin B, Holinski-Feder E, Lochmüller H, Horvath R. *Neuromuscul Disord*. 2010;20(11):720-724. https://doi.org/10.1016/j.nmd.2010.06.017

Late-onset ptosis and myopathy in a patient with a heterozygous insertion in POLG2. Walter MC, Czermin B, Muller-Ziermann S, Bulst S, Stewart JD, Hudson G, Schneiderat P, **Abicht A**, Holinski-Feder E, Lochmüller H, Chinnery PF, Klopstock T, Horvath R. *J Neurol*. 2010;257(9):1517-1523. https://doi.org/10.1007/s00415-010-5565-9

Molecular characterisation of congenital myasthenic syndromes in Southern Brazil. Mihaylova V, Scola RH, Gervini B, Lorenzoni PJ, Kay CK, Werneck LC, Stucka R, Guergueltcheva V, von der Hagen M, Huebner A, **Abicht A**, Müller JS, Lochmüller H. *J Neurol Neurosurg Psychiatry*. 2010;81(9):973-977. https://doi.org/10.1136/jnnp.2009.177816

Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations. Schara U, Christen H-J, Durmus H, Hietala M, Krabetz K, Rodolico C, Schreiber G, Topaloglu H, Talim B, Voss W, Pihko H, **Abicht A**, Müller JS, Lochmüller H. *Eur J Paediatr Neurol*. 2010;14(4):326-333. https://doi.org/10.1016/j.ejpn.2009.09.009

2009

Severe Myoclonic Epilepsy in Infancy - Adult Phenotype with Bradykinesia, Hypomimia, and Perseverative Behavior: Report of Five Cases. Martin P, Rautenstrauß B, **Abicht A**, Fahrbach J, Koster S. *Mol Syndromol*. 2010;1(5):231-238. https://doi.org/10.1159/000326746

Refinement of the clinical phenotype in musk-related congenital myasthenic syndromes. Mihaylova V, Salih MAM, Mukhtar MM, Abuzeid HA, El-Sadig SM, von der Hagen M, Huebner A, Nürnberg G, **Abicht A**, Müller JS, Lochmüller H, Guergueltcheva V. *Neurology*. 2009;73(22):1926-1928. https://doi.org/10.1212/WNL.0b013e3181c3fce9

Ephedrine therapy in eight patients with congenital myasthenic syndrome due to DOK7 mutations. Schara U, Barisic N, Deschauer M, Lindberg C, Straub V, Strigl-Pill N, Wendt M, **Abicht A**, Müller JS, Lochmüller H. *Neuromuscul Disord*. 2009;19(12):828-832. https://doi.org/10.1016/j.nmd.2009.09.008

In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes. Bulst S, **Abicht A**, Holinski-Feder E, Müller-Ziermann S, Koehler U, Thirion C, Walter MC, Stewart JD, Chinnery PF, Lochmüller H, Horvath R. *Hum Mol Genet*. 2009;18(9):1590-1599. https://doi.org/10.1093/hmg/ddp074

Heteroplasmic mutation in the anticodon-stem of mitochondrial tRNA(Val) causing MNGIE-like gastrointestinal dysmotility and cachexia. Horváth R, Bender A, **Abicht A**, Holinski-Feder E, Czermin B, Trips T, Schneiderat P, Lochmüller H, Klopstock T. *J Neurol*. 2009;256(5):810-815. https://doi.org/10.1007/s00415-009-5023-8

Mitochondrial neurogastrointestinal encephalomyopathy mimicking anorexia nervosa. Feddersen B, DE LA Fontaine L, Sass JO, Lutz J, **Abicht A**, Klopstock T, Verma IC, Meisenzahl E, Pogarell O. *Am J Psychiatry*. 2009;166(4):494-495. https://doi.org/10.1176/appi.ajp.2008.08101525

2008

Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. Mihaylova V, Müller JS, Vilchez JJ, Salih MA, Kabiraj MM, D'Amico A, Bertini E, Wölfle J, Schreiner F, Kurlemann G, Rasic VM, Siskova D, Colomer J, Herczegfalvi A, Fabriciova K, Weschke B, Scola R, Hoellen F, Schara U, **Abicht A**, Lochmüller H. *Brain*. 2008;131(Pt 3):747-759. https://doi.org/10.1093/brain/awm325

2007

Congenital myasthenic syndromes: spotlight on genetic defects of neuromuscular transmission. Müller JS, Mihaylova V, **Abicht A**, Lochmüller H. *Expert Rev Mol Med*. 2007;9(22):1-20. https://doi.org/10.1017/S1462399407000427

Phenotypical spectrum of DOK7 mutations in congenital myasthenic syndromes. Müller JS, Herczegfalvi A, Vilchez JJ, Colomer J, Bachinski LL, Mihaylova V, Santos M, Schara U, Deschauer M, Shevell M, Poulin C, Dias A, Soudo A, Hietala M, Aärimaa T, Krahe R, Karcagi V, Huebner A, Beeson D, **Abicht A**, Lochmüller H. *Brain*. 2007;130(Pt 6):1497-1506. https://doi.org/10.1093/brain/awm068

2006

Impaired receptor clustering in congenital myasthenic syndrome with novel RAPSN mutations. Müller JS, Baumeister SK, Rasic VM, Krause S, Todorovic S, Kugler K, Müller-Felber W, **Abicht A**, Lochmüller H. *Neurology*. 2006;67(7):1159-1164. https://doi.org/10.1212/01.wnl.0000233837.79459.40

CHRND mutation causes a congenital myasthenic syndrome by impairing co-clustering of the acetylcholine receptor with rapsyn. Müller JS, Baumeister SK, Schara U, Cossins J, Krause S, von der Hagen M, Huebner A, Webster R, Beeson D, Lochmüller H, **Abicht A**. *Brain*. 2006;129(Pt 10):2784-2793. https://doi.org/10.1093/brain/awl188

Long-term improvement of slow-channel congenital myasthenic syndrome with fluoxetine. Colomer J, Müller JS, Vernet A, Nascimento A, Pons M, Gonzalez V, **Abicht A**, Lochmüller H. *Neuromuscul Disord*. 2006;16(5):329-333. https://doi.org/10.1016/j.nmd.2006.02.009

Facing the genetic heterogeneity in neuromuscular disorders: linkage analysis as an economic diagnostic approach towards the molecular diagnosis. von der Hagen M, Schallner J, Kaindl AM, Koehler K, Mitzscherling P, **Abicht A**, Grieben U, Korinthenberg R, Kress W, von Moers A, Müller JS, Schara U, Vorgerd M, Walter MC, Müller-Reible C, Hübner C, Lochmüller H, Huebner A. *Neuromuscul Disord*. 2006;16(1):4-13. https://doi.org/10.1016/j.nmd.2005.10.001

Leigh syndrome caused by mutations in the flavoprotein (Fp) subunit of succinate dehydrogenase (SDHA). Horváth R, **Abicht A**, Holinski-Feder E, Laner A, Gempel K, Prokisch H, Lochmüller H, Klopstock T, Jaksch M. *J Neurol Neurosurg Psychiatry*. 2006;77(1):74-76. https://doi.org/10.1136/jnnp.2005.067041

2005

An intronic base alteration of the CHRNE gene leading to a congenital myasthenic syndrome. Müller JS, Stucka R, Neudecker S, Zierz S, Schmidt C, Huebner A, Lochmüller H, **Abicht A**. *Neurology*. 2005;65(3):463-465. https://doi.org/10.1212/01.wnl.0000172346.26219.fd

An Iranian family with congenital myasthenic syndrome caused by a novel acetylcholine receptor mutation (CHRNE K171X). Soltanzadeh P, Müller JS, Ghorbani A, **Abicht A**, Lochmüller H, Soltanzadeh A. *J Neurol Neurosurg Psychiatry*. 2005;76(7):1039-1040. https://doi.org/10.1136/jnnp.2004.059436

Clinical variability of CMS-EA (congenital myasthenic syndrome with episodic apnea) due to identical CHAT mutations in two infants. Barisic N, Müller JS, Paucic-Kirincic E, Gazdik M, Lah-Tomulic K, Pertl A, Sertic J, Zurak N, Lochmüller H, **Abicht A**. *Eur J Paediatr Neurol*. 2005;9(1):7-12. https://doi.org/10.1016/j.ejpn.2004.10.008

2004

A newly identified chromosomal microdeletion of the rapsyn gene causes a congenital myasthenic syndrome. Müller JS, **Abicht A**, Christen H-J, Stucka R, Schara U, Mortier W, Huebner A, Lochmüller H. *Neuromuscul Disord*. 2004;14(11):744-749. https://doi.org/10.1016/j.nmd.2004.06.010

Mutation history of the roma/gypsies. Morar B, Gresham D, Angelicheva D, Tournev I, Gooding R, Guergueltcheva V, Schmidt C, **Abicht A**, Lochmuller H, Tordai A, Kalmar L, Nagy M, Karcagi V, Jeanpierre M, Herczegfalvi A, Beeson D, Venkataraman V, Warwick Carter K, Reeve J, de Pablo R, Kucinskas V, Kalaydjieva L. *Am J Hum Genet*. 2004;75(4):596-609. https://doi.org/10.1086/424759

The congenital myasthenic syndrome mutation RAPSN N88K derives from an ancient Indo-European founder. Müller JS, **Abicht A**, Burke G, Cossins J, Richard P, Baumeister SK, Stucka R, Eymard B, Hantaï D, Beeson D, Lochmüller H. *J Med Genet*. 2004;41(8):e104. https://doi.org/10.1136/jmg.2004.021139

Synaptic congenital myasthenic syndrome in three patients due to a novel missense mutation (T441A) of the COLQ gene. Müller JS, Petrova S, Kiefer R, Stucka R, König C, Baumeister SK, Huebner A, Lochmüller H, **Abicht A**. *Neuropediatrics*. 2004;35(3):183-189. https://doi.org/10.1055/s-2004-820996

2003

Rapsyn N88K is a frequent cause of congenital myasthenic syndromes in European patients. Müller JS, Mildner G, Müller-Felber W, Schara U, Krampfl K, Petersen B, Petrova S, Stucka R, Mortier W, Bufler J, Kurlemann G, Huebner A, Merlini L, Lochmüller H, **Abicht A**. *Neurology*. 2003;60(11):1805-1810. https://doi.org/10.1212/01.wnl.0000072262.14931.80

Congenital myasthenic syndrome due to a novel missense mutation in the gene encoding choline acetyltransferase. Schmidt C, **Abicht A**, Krampfl K, Voss W, Stucka R, Mildner G, Petrova S, Schara U, Mortier W, Bufler J, Huebner A, Lochmüller H. *Neuromuscul Disord*. 2003;13(3):245-251. https://doi.org/10.1016/s0960-8966(02)00273-0

2002

What's in the serum of seronegative MG and LEMS?: MuSK et al. Abicht A, Lochmüller H. *Neurology*. 2002;59(11):1672-1673. https://doi.org/10.1212/01.wnl.0000041026.90947.79

Congenital myasthenic syndrome (CMS) in three European kinships due to a novel splice mutation (IVS7 -2 A/G) in the epsilon acetylcholine receptor (AChR) subunit gene. Barisic N, Schmidt C, Sidorova OP, Herczegfalvi A, Gekht BM, Song I-H, Stucka R, Karcagi V, **Abicht A**, Lochmüller H. *Neuropediatrics*. 2002;33(5):249-254. https://doi.org/10.1055/s-2002-36738

A newly identified chromosomal microdeletion and an N-box mutation of the AChR epsilon gene cause a congenital myasthenic syndrome. Abicht A, Stucka R, Schmidt C, Briguet A, Höpfner S, Song I-H, Pongratz D, Müller-Felber W, Ruegg MA, Lochmüller H. *Brain*. 2002;125(Pt 5):1005-1013. https://doi.org/10.1093/brain/awf095

2001

Deactivation and desensitization of mouse embryonic- and adult-type nicotinic receptor channel currents. Jahn K, Mohammadi B, Krampfl K, **Abicht A**, Lochmüller H, Bufler J. *Neurosci Lett*. 2001;307(2):89-92. https://doi.org/10.1016/s0304-3940(01)01929-2

2000

Technology evaluation: edrecolomab, Centocor Inc. Abicht A, Lochmüller H. *Curr Opin Mol Ther*. 2000;2(5):593-600.

A modified alignment of human and rodent 5' untranslated sequences of the acetylcholine receptor epsilon subunit gene reveals additional regions of high homology. Stucka R, **Abicht A**, Song IH, Bönsch D, Deufel T, Lochmüller H. *Neuromuscul Disord*. 2000;10(3):213-214. https://doi.org/10.1016/s0960-8966(99)00112-1

Leber's hereditary optic neuropathy presenting as multiple sclerosis-like disease of the CNS. Horváth R, **Abicht A**, Shoubridge EA, Karcagi V, Rózsa C, Komoly S, Lochmüller H. *J Neurol*. 2000;247(1):65-67. https://doi.org/10.1007/s004150050015

1999

A common mutation (epsilon1267delG) in congenital myasthenic patients of Gypsy ethnic origin. Abicht A, Stucka R, Karcagi V, Herczegfalvi A, Horváth R, Mortier W, Schara U, Ramaekers V, Jost W, Brunner J, Janssen G, Seidel U, Schlotter B, Müller-Felber W, Pongratz D, Rüdel R, Lochmüller H. *Neurology*. 1999;53(7):1564-1569. https://doi.org/10.1212/wnl.53.7.1564

Technology evaluation: CRIB (CNTF delivery) CytoTherapeutics Inc. Abicht A, Lochmüller H. *Curr Opin Mol Ther*. 1999;1(5):645-650.

Localization of transforming growth factor beta in association with neuromuscular junctions in adult human muscle. Toepfer M, Fischer P, **Abicht A**, Lochmüller H, Pongratz D, Müller-Felber W. *Cell Mol Neurobiol*. 1999;19(2):297-300. https://doi.org/10.1023/a:1006989530148

1998

Myotonic ADR-MDX mutant mice show less severe muscular dystrophy than MDX mice. Krämer R, Lochmüller H, **Abicht A**, Rüdel R, Brinkmeier H. *Neuromuscul Disord*. 1998;8(8):542-550. https://doi.org/10.1016/s0960-8966(98)00078-9

1997

Congenital myasthenic syndromes: clinical and genetic analysis of 18 patients. Abicht A, Müller-Felber W, Fischer P, Jakob I, Kürz L, Rudel R, Mortier W, Pongratz D, Lochmüller H. *Eur J Med Res*. 1997;2(12):515-522.

1993

Congenital Myasthenic Syndromes. Abicht A, Müller JS, Lochmüller H. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJ, Stephens K, Amemiya A, eds. *GeneReviews(®)*. University of Washington, Seattle; 1993.