Veröffentlichungen der BMBF-geförderten Forschungsverbünde für seltene Erkrankungen, 2020

31. Dezember 2020 (CONNECT-GENERATE)
Autoimmune encephalitis: novel therapeutic targets at the preclinical level
Expert Opin Ther Targets. 2020 Dec 31;1-11 [Epub ahead of print] >>PubMed-Link<<
Josefine Sell, Holger Haselmann, Stefan Hallermann, Michael Hust, Christian Geis

21. Dezember 2020 (MitoNet)
Expanding the clinical and genetic spectrum of FDXR deficiency by functional validation of variants of uncertain significance
Hum Mutat. 2020 Dec 21. [Epub ahead of print] >>PubMed-Link<<
Sarah L Stenton, Dorota Piekutowska-Abramczuk, Lea Kulterer, Robert Kopajtich, Kristl G Claeys, Elżbieta Ciara, Johannes Eisen, Rafał Płoski, Ewa Pronicka, Katarzyna Malczyk, Matias Wagner, Saskia B Wortmann, Holger Prokisch

18. Dezember 2020 (GeNeRARe)
Abnormalities of synaptic mitochondria in autism spectrum disorder and related neurodevelopmental disorders
J Mol Med. 2020 Dec 18. [Epub ahead of print] >>PubMed-Link<<
Liliana Rojas-Charry, Leonardo Nardi, Axel Methner, Michael J. Schmeisser

15. Dezember 2020 (CONNECT-GENERATE)
Low-Avidity CD4 + T Cell Responses to SARS-CoV-2 in Unexposed Individuals and Humans with Severe COVID-19
Immunity. 2020 Dec 15 >>PubMed-Link<<
Petra Bacher, Elisa Rosati, Daniela Esser, Gabriela Rios Martini, Carina Saggau, Esther Schiminsky, Justina Dargvainiene, Ina Schröder, Imke Wieters, Yascha Khodamoradi, Fabian Eberhardt, Maria J G T Vehreschild, Holger Neb, Michael Sonntagbauer, Claudio Conrad, Florian Tran, Philip Rosenstiel, Robert Markewitz, Klaus-Peter Wandinger, Max Augustin, Jan Rybniker, Matthias Kochanek, Frank Leypoldt, Oliver A Cornely, Philipp Koehler, Andre Franke, Alexander Scheffold

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Bilateral visual improvement with unilateral gene therapy injection for Leber hereditary optic neuropathy
Sci Transl Med. 2020 Dec 9. [Epub ahead of print] *>>PubMed-Link<<*
Patrick Yu-Wai-Man, Nancy J Newman, Valerio Carelli, Mark L Moster, Valerie Biousse, Alfredo A Sadun, Thomas Klopstock, Catherine Vignal-Clermont, Robert C Sergott, Günther Rudolph, Chiara La Morgia, Rustum Karanjia, Magali Taiel, Laure Blouin, Pierre Burguière, Gerard Smits, Caroline Chevalier, Harvey Masonson, Yordak Salermo, Barrett Katz, Serge Picaud, David J Calkins, José-Alain Sahel

7. Dezember 2020 (GeNeRARe)
Aplasia cutis congenita in a CDC42-related developmental phenotype
Am J Med Genet A. 2020 Dec 7. [Epub ahead of print] *>>PubMed-Link<<*
Franziska Schnabel, Susanne B Kamphausen, Rudolf Funke, Silke Kaulfuß, Bernd Wollnik, Martin Zenker

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Dynamics of salivary markers of kidney functions in acute and chronic kidney diseases
Sci Rep. 2020 Dec 4.[Epub ahead of print] *>>PubMed-Link<<*
Alexandra Gaál Kovalčíková, Kristína Pavlov, Róbert Lipták, Marianna Hladová, Emese Renczés, Peter Boor, Ľudmila Podracká, Katarína Šebeková, Július Hodosy, Ľubomíra Tóthová, Peter Celec

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Extended clinical and immunological phenotype and transplant outcome in CD27 and CD70 deficiency
Blood. 2020 Dec 3 >>PubMed-Link<<
Sujal Ghosh, Sevgi Köstel Bal, Emily S J Edwards, Bethany Pillay, Raúl Jiménez Heredia, Funda Erol Cipe , Geetha Rao , Elisabeth Salzer, Samaneh Zoghi, Hassan Abolhassani, Tooba Momen, Emma Gostick, David A Price, Yu Zhang, Andrew J Oler, Claudia Gonzaga-Jauregui, Baran Erman, Ayse Metin, Inci Ilhan, Sule Haskologlu, Candan Islamoglu, Kubra Baskin, Serdar Ceylaner, Ebru Yilmaz, Ekrem Unal, Musa Karakukcu, Dagmar Berghuis, Theresa Cole, Aditya K Gupta, Fabian Hauck, Hubert Kogler, Andy I M Hoepelman, Safa Baris, Elif Karakoc-Aydiner,

Ahmet Ozen, Leo Kager, Dirk Holzinger, Michael Paulussen, Renate Krüger, Roland Meisel, Prasad T Oommen, Emma Morris, Benedicte Neven, Austen Worth, Joris van Montfrans, Pieter L A Fraaij, Sharon Choo, Figen Dogu, E Graham Davies, Siobhan Burns, Gregor Dückers, Ruy Perez Becker, Horst von Bernuth, Sylvain Latour, Maura Faraci, Marco Gattorno, Helen C Su, Qiang Pan-Hammarström, Lennart Hammarström, Michael J Lenardo, Cindy S Ma, Tim Niehues, Asghar Aghamohammadi, Nima Rezaei, Aydan Ikinciogullari, Stuart G Tangye, Arjan C Lankester, Kaan Boztug

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Generation of the CRISPR/Cas9-mediated KIF1C knock-out human iPSC line HIHRSi003-A-1
Stem Cell Research. 2020 Dec. *>>PubMed-Link<<*
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A reciprocal regulation of spermidine and autophagy in podocytes maintains the filtration barrier
Kidney Int. 2020 Dec. Epub 2020 Jun 27. >>PubMed-Link<<
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Transdiagnostic hippocampal damage patterns in neuroimmunological disorders
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The Dimensions of Primary Mitochondrial Disorders
Front Cell Dev Biol. 2020 Nov 26. [Epub ahead of print] *>>PubMed-Link<<*
Lea D Schlieben, Holger Prokisch

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Recurrent acute liver failure in alanyl-tRNA synthetase-1 (AARS1) deficiency
Mol Genet Metab Rep. 2020 Nov 22. >>PubMed-Link<<
Lara M Marten, Florian Brinkert, Desirée E C Smith, Holger Prokisch, Maja Hempel, René Santer

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Vascular neutrophilic inflammation and immunothrombosis distinguish severe COVID-19 from influenza pneumonia
J Thromb Haemost. 2020 Nov 20 [Epub ahead of print] >>PubMed-Link<<
Leo Nicolai, Alexander Leunig, Sophia Brambs, Rainer Kaiser, Markus Joppich, Marie-Louise Hoffknecht, Christoph Gold, Anouk Engel, Vivien Polewka, Maximilian Muenchhoff, Johannes C Hellmuth, Adrian Ruhle, Stephan Ledderose, Tobias Weinberger, Heiko Schulz, Clemens Scherer, Martina Rudelius, Michael Zoller, Oliver T Keppler, Bernhard Zwißler, Michael von Bergwelt-Baildon, Stefan Kääb, Ralf Zimmer, Roman D Bülow, Saskia von Stillfried, Peter Boor, Steffen Massberg, Kami Pekayvaz, Konstantin Stark

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PLoS One. 2020 Nov 13. [Epub ahead of print] *>>PubMed-Link<<*
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A Therapeutic Non-self-reactive SARS-CoV-2 Antibody Protects from Lung Pathology in a COVID-19 Hamster Model
Cell. 2020 Nov 12. [Epub ahead of print] *>>PubMed-Link<<*
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Endres, Dietmar Schmitz, Lara Maria Jeworowski, Anja Richter, Marie Luisa Schmidt, Tatjana Schwarz, Marcel Alexander Müller, Christian Drosten, Daniel Wendisch, Leif E Sander, Nikolaus Osterrieder, Ian A Wilson, Harald Prüss

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Occurrence of Portal Hypertension and Its Clinical Course in Patients With Molecularly Confirmed Autosomal Recessive Polycystic Kidney Disease (ARPKD)
Front Pediatr. 2020 Nov 12. [Epub ahead of print] *>>PubMed-Link<<*
Dorota Wicher, Ryszard Grenda, Mikołaj Teisseyre, Marek Szymczak, Paulina Halat-Wolska, Dorota Jurkiewicz, Max Christoph Liebau, Elżbieta Ciara, Małgorzata Rydzanicz, Joanna Kosińska, Krystyna Chrzanowska, Irena Jankowska

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Intracellular A53T Mutant α-Synuclein Impairs Adult Hippocampal Newborn Neuron Integration
Front Cell Dev Biol. 2020 Nov 11. [Epub ahead of print] *>>PubMed-Link<<*
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Paroxysmal and non-paroxysmal dystonia in 3 patients with biallelic ECHS1 variants: Expanding the neurological spectrum and therapeutic approaches
Eur J Med Genet. 2020 Nov. [Epub ahead of print] *>>PubMed-Link<<*
Sabine Illsinger, G Christoph Korenke, Sylvia Boesch, Michael Nocker, Daniela Karall, Jean M Nuoffer, Lucia Laugwitz, Johannes A Mayr, Sabine Scholl-Bürgi, Peter Freisinger, Tobias Kowald, Stefan Kölker, Holger Prokisch, Tobias B Haack

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Clinical Relevance of Standardized Mobile Gait Tests. Reliability Analysis Between Gait Recordings at Hospital and Home in Parkinson's Disease: A Pilot Study
J Parkinsons Dis. 2020. [Epub ahead of print] *>>PubMed-Link<<*
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The carboxy-terminus of the human ARPKD protein fibrocystin can control STAT3 signalling by regulating SRC-activation
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Claudia Dafinger, Amrei M Mandel, Alina Braun, Heike Göbel, Kathrin Burgmaier, Laura Massella, Antonio Mastrangelo, Jörg Dötsch, Thomas Benzing, Thomas Weimbs, Bernhard Schermer, Max C Liebau

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Front Pediatr. 2020 Oct 23. *>>PubMed-Link<<*
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Early-onset autoimmunity associated with SOCS1 haploinsufficiency
Nat Commun. 2020 Oct 21 >>PubMed-Link<<
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The clinical significance ofA2ML1variants in Noonan syndrome has to be reconsidered
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Prolonged podocyte depletion in larval zebrafish resembles mammalian focal and segmental glomerulosclerosis
FASEB J. 2020 Oct 18. [Epub ahead of print] *>>PubMed-Link<<*
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13. Oktober 2020 (MitoNet)
Biotinidase deficiency: A treatable cause of hereditary spastic paraparesis
Neurol Genet. 2020 Oct 13. eCollection 2020 Dec. >>PubMed-Link<<
Florentine Radelfahr, Korbinian M Riedhammer, Leonie F Keidel, Gwendolyn Gramer, Thomas Meitinger, Thomas Klopstock, Matias Wagner

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CHIP mutations affect the heat shock response differently in human fibroblasts and iPSC-derived neurons
Dis Model Mech. 2020 Oct 12. *>>PubMed-Link<<*
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High frequency of variants in genes associated with primary immunodeficiencies in patients with rheumatic diseases with secondary hypogammaglobulinaemia
Ann Rheum Dis. 2020 Oct 12. [Epub ahead of print] *>>PubMed-Link<<*
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Matched Family Donor Lymphocyte Infusions as First Cellular Therapy for Patients with Severe Primary T Cell Deficiencies
Biol Blood Marrow Transplant. 2020 Oct 3 [Epub ahead of print] >>PubMed-Link<<
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The proteomic landscape of small urinary extracellular vesicles during kidney transplantation
J Extracell Vesicles. 2020 Oct.[Epub ahead of print] *>>PubMed-Link<<*
Fabian Braun, Markus Rinschen, Denise Buchner, Katrin Bohl, Ingo Plagmann, Daniel Bachurski, Martin Richard Späth, Philipp Antczak, Heike Göbel, Corinna Klein, Jan-Wilm Lackmann, Oliver Kretz, Victor G Puelles, Roger Wahba, Michael Hallek, Bernhard Schermer, Thomas Benzing, Tobias B Huber, Andreas Beyer, Dirk Stippel, Christine E Kurschat, Roman-Ulrich Müller

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Macrophage frequency in the bone marrow correlates with morphologic subtype of myeloproliferative neoplasm
Ann Hematol. 2020 Oct 26. [Epub ahead of print] *>>PubMed-Link<<*
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Novel nephronophthisis-associated variants reveal functional importance of MAPKBP1 dimerization for centriolar recruitment
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MIC26 and MIC27 cooperate to regulate cardiolipin levels and the landscape of OXPHOS complexes
Life Sci Alliance. 2020 Oct 1. [Epub ahead of print] *>>PubMed-Link<<*
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29. September 2020 (NEOCYST)
Severe neurological outcomes after very early bilateral nephrectomies in patients with autosomal recessive polycystic kidney disease (ARPKD)
Sci Rep. 2020 Sep 29. [Epub ahead of print] *>>PubMed-Link<<*
Kathrin Burgmaier, Gema Ariceta, Martin Bald, Anja Katrin Buescher, Mathias Burgmaier, Florian Erger, Michaela Gessner, Ibrahim Gokce, Jens König, Claudia Kowalewska, Laura Massella, Antonio Mastrangelo, Djalila Mekahli, Lars Pape, Ludwig Patzer, Alexandra Potemkina, Gesa Schalk, Raphael Schild, Rukshana Shroff, Maria Szczepanska, Katarzyna Taranta-Janusz, Marcin Tkaczyk, Lutz Thorsten Weber, Elke Wühl, Donald Wurm, Simone Wygoda, Ilona Zagozdzon, Jörg Dötsch, Jun Oh, Franz Schaefer, Max Christoph Liebau

24. September 2020 (MitoNet)
Bi-allelic pathogenic variants in NDUFC2 cause early-onset Leigh syndrome and stalled biogenesis of complex I
EMBO Mol Med. 2020 Sep 24. [Epub ahead of print] *>>PubMed-Link<<*
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Systematic evaluation of olfaction in patients with hereditary cystic kidney diseases/renal ciliopathies
J Med Genet. 2020 Sep 11. [Epub ahead of print] *>>PubMed-Link<<*
Mareike Dahmer-Heath, Valentin Schriever, Sabine Kollmann, Carolin Schleithoff, Andrea Titieni, Metin Cetiner, Ludwig Patzer, Burkhard Tönshoff, Matthias Hansen, Petra Pennekamp, Joachim Gerß, Martin Konrad, Jens König

3. September 2020 (CONNECT-GENERATE)
Cerebrospinal fluid findings in patients with myelin oligodendrocyte glycoprotein (MOG) antibodies. Part 2: Results from 108 lumbar punctures in 80 pediatric patients
J Neuroinflammation. 2020 Sep 3. [Epub ahead of print] *>>PubMed-Link<<*
Sven Jarius, Christian Lechner, Eva M Wendel, Matthias Baumann, Markus Breu, Mareike Schimmel, Michael Karenfort, Adela Della Marina, Andreas Merkenschlager, Charlotte Thiels, Astrid Blaschek, Michela Salandin, Steffen Leiz, Frank Leypoldt, Alexander Pschibul, Annette Hackenberg, Andreas Hahn, Steffen Syrbe, Jurgis Strautmanis, Martin Häusler, Peter Krieg, Astrid Eisenkölbl, Johannes Stoffels, Matthias Eckenweiler, Ilya Ayzenberg, Jürgen Haas, Romana Höftberger, Ingo Kleiter, Mirjam Korporal-Kuhnke, Marius Ringelstein, Klemens Ruprecht, Nadja Siebert, Kathrin Schanda, Orhan Aktas, Friedemann Paul, Markus Reindl, Brigitte Wildemann, Kevin Rostásy, in cooperation with the BIOMARKER study group and the Neuromyelitis optica Study Group (NEMOS)

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Cerebrospinal fluid findings in patients with myelin oligodendrocyte glycoprotein (MOG) antibodies. Part 1: Results from 163 lumbar punctures in 100 adult patients
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Cognitive impact of neuronal antibodies: encephalitis and beyond
Transl Psychiatry. 2020 Sep 1. [Epub ahead of print] *>>PubMed-Link<<*
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Anti-tumor mechanisms of rigosertib in colorectal cancer
Ann Oncol. 2020 Sep 01. [Epub ahead of print]
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Distinct Modes of Balancing Glomerular Cell Proteostasis in Mucolipidosis Type II and III Prevent Proteinuria
J Am Soc Nephrol. 2020 Aug. [Epub ahead of print] *>>PubMed-Link<<*
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29. August 2020 (TreatHSP.net)
Long-term course of anterior spinal cord herniation presenting with an upper motor neuron syndrome: case report illustrating diagnostic and therapeutic implications
BMC Neurol. 2020 Aug 29. *>>PubMed-Link<<*
Martin Regensburger, Johannes C M Schlachetzki, Jörg Klekamp, Arnd Doerfler, Jürgen Winkler

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Mitochondrial Regulation of the 26S Proteasome
Cell Rep. 2020 Aug 25. [Epub ahead of print] *>>PubMed-Link<<*
Thomas Meul, Korbinian Berschneider, Sabine Schmitt, Christoph H Mayr, Laura F Mattner, Herbert B Schiller, Ayse S Yazgili, Xinyuan Wang, Christina Lukas, Camille Schlesser, Cornelia Prehn, Jerzy Adamski, Elisabeth Graf, Thomas Schwarzmayr, Fabiana Perocchi, Alexandra Kukat, Aleksandra Trifunovic, Laura Kremer, Holger Prokisch, Bastian Popper, Christine von Toerne, Stefanie M Hauck, Hans Zischka, Silke Meiners

25. August 2020 (CONNECT-GENERATE)
Gender issues of antibody-mediated diseases in neurology: (NMOSD/autoimmune encephalitis/MG)
Ther Adv Neurol Disord. 2020 Aug 25. *>>PubMed-Link<<*
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24. August 2020 (NEOCYST)
Metabolic Changes in Polycystic Kidney Disease as a Potential Target for Systemic Treatment
Int J Mol Sci. 2020 Aug 24. [Epub ahead of print] *>>PubMed-Link<<*
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Orthotopic Kidney Auto-Transplantation in a Porcine Model Using 24 Hours Organ Preservation And Continuous Telemetry
J Vis Exp. 2020 Aug 21. [Epub ahead of print] *>>PubMed-Link<<*
Wen-Jia Liu, Lisa Ernst, Benedict Doorschodt, Jan Bednarsch, Felix Becker, Richi Nakatake, Yuki Masano, Ulf Peter Neumann, Sven Arke Lang, Peter Boor, Isabella Lurje, Georg Lurje, René Tolba, Zoltan Czigany

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SARS-CoV-2 renal tropism associates with acute kidney injury
Lancet. 2020 Aug 14. [Epub ahead of print] *>>PubMed-Link<<*
Fabian Braun, Marc Lütgehetmann, Susanne Pfefferle, Milagros N Wong, Alexander Carsten, Maja T Lindenmeyer, Dominik Nörz, Fabian Heinrich, Kira Meißner, Dominic Wichmann, Stefan Kluge, Oliver Gross, Klaus Pueschel, Ann S Schröder, Carolin Edler, Martin Aepfelbacher, Victor G Puelles, Tobias B Huber

12. August 2020 (Treat-ION)
Predicting functional effects of missense variants in voltage-gated sodium and calcium channels
Sci Transl Med. 2020 Aug 12. [Epub ahead of print] *>>PubMed-Link<<*
Henrike O Heyne, David Baez-Nieto, Sumaiya Iqbal, Duncan S Palmer, Andreas Brunklaus, Patrick May, Epi25 Collaborative; Katrine M Johannesen, Stephan Lauxmann, Johannes R Lemke, Rikke S Møller, Eduardo Pérez-Palma, Ute I Scholl, Steffen Syrbe, Holger Lerche, Dennis Lal, Arthur J Campbell, Hao-Ran Wang, Jen Pan, Mark J Daly

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Variants of SOS2 are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications
Eur J Hum Genet. 2020 Aug 12. [Epub ahead of print] *>>PubMed-Link<<*
Christina Lissewski, Valérie Chune, Francesca Pantaleoni, Alessandro De Luca, Yline Capri, Julia Brinkmann, Francesca Lepri, Paola Daniele, Erika Leenders, Laura Mazzanti, Emanuela Scarano, Francesca Clementina Radio, Kerstin Kutsche, Alma Kuechler, Marion Gérard, Kara Ranguin, Marine Legendre, Yoann Vial, Ineke van der Burgt, Tuula Rinne, Elena Andreucci, Gioia Mastromoro, Maria Cristina Digilio, Hélène Cave, Marco Tartaglia, Martin Zenker

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Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia
Am J Hum Genet. 2020 Aug 6. [Epub ahead of print] *>>PubMed-Link<<*
Ralf A Husain, Mona Grimmel, Matias Wagner, J Christopher Hennings, Christian Marx, René G Feichtinger, Abdelkrim Saadi, Kevin Rostásy, Florentine Radelfahr, Andrea Bevot, Marion Döbler-Neumann, Hans Hartmann, Laurence Colleaux, Isabell Cordts, Xenia Kobeleva, Hossein Darvish, Somayeh Bakhtiari, Michael C Kruer, Arnaud Besse, Andy Cheuk-Him Ng, Diana Chiang, Francois Bolduc, Abbas Tafakhori, Shrikant Mane, Saghar Ghasemi Firouzabadi, Antje K Huebner, Rebecca Buchert, Stefanie Beck-Woedl, Amelie J Müller, Lucia Laugwitz, Thomas Nägele, Zhao-Qi Wang, Tim M Strom, Marc Sturm, Thomas Meitinger, Thomas Klockgether, Olaf Riess, Thomas Klopstock, Ulrich Brandl, Christian A Hübner, Marcus Deschauer, Johannes A Mayr, Penelope E Bonnen, Ingeborg Krägeloh-Mann, Saskia B Wortmann, Tobias B Haack
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Functional consequences of SLC1A3 mutations associated with episodic ataxia 6
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A Salvage Pathway Maintains Highly Functional Respiratory Complex I
Nat Commun. 2020 Apr 2. *>>PubMed-Link<<*
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Nisha Verwey, Isabella Gazzoli, Sabine Krause, Kamel Mamchaoui, Vincent Mouly, Annemieke Aartsma-Rus

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Inhibition of Activin/Myostatin signalling induces skeletal muscle hypertrophy but impairs mouse testicular development
Eur J Transl Myol. 2020 Apr 1. *>>PubMed-Link<<*
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17. März 2020 (Treat-ION)
Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders
Genome Med. 2020 Mar 17. *>>PubMed-Link<<*
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Dysregulated mesenchymal PDGFR-β drives kidney fibrosis
EMBO Mol Med. 2020 März 6 *>>PubMed-Link<<*
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Increased glutamate transporter-associated anion currents cause glial apoptosis in episodic ataxia 6
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Structural Noninfectious Manifestations of the Central Nervous System in Common Variable Immunodeficiency Disorders
J Allergy Clin Immunol Pract. 2020 Mar. [Epub ahead of print] *>>PubMed-Link<<*
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24. Februar 2020 (STOP-FSGS)
Crystal Clots as Therapeutic Target in Cholesterol Crystal Embolism
Circ Res. 2020 Feb 24. [Epub ahead of print] ***>>PubMed-Link<<***
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J Am Soc Nephrol. 2020 Feb 21. [Epub ahead of print] ***>>PubMed-Link<<***
Barbara Mara Klinkhammer, Sonja Djudjaj, Uta Kunter, Runolfur Palsson, Vidar Orn Edvardsson, Thorsten Wiech, Margret Thorsteinsdottir, Sverrir Hardarson, Orestes Foresto-Neto, Shrikant R. Mulay, Marcus Johannes Moeller, Wilhelm Jahnen-Dechent, Jürgen Floege, Hans-Joachim Anders and Peter Boor

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Respiratory chain signalling is essential for adaptive remodelling following cardiac ischaemia
J Cell Mol Med. 2020 Feb 10. [Epub ahead of print]*>>PubMed-Link<<*
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Bioenergetic consequences from xenotopic expression of a tunicate AOX in mouse mitochondria: Switch from RET and ROS to FET

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Neurol Genet. 2020 Feb. [Epub ahead of print] *>>PubMed-Link<<*
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Functional characterization of the first missense variant in CEP78, a founder allele associated with cone-rod dystrophy, hearing loss, and reduced male fertility
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J Clin Immunol. 2020 Jan 24 [Epub ahead of print] ***>>PubMed-Link<<***
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Modeling Cell-Cell Interactions in Parkinson's Disease Using Human Stem Cell-Based Models
Front Cell Neurosci. 2020 Jan 17. [Epub ahead of print] *>>PubMed-Link<<*
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Delineating MT-ATP6-associated disease: From isolated neuropathy to early onset neurodegeneration.
Neurol Genet. 2020 Jan 13 *>> PubMed-Link<<*
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Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency
Am J Hum Genet. 2020 Jan 2. [Epub ahead of print] *>> PubMed-Link<<*
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The diagnosis of inborn errors of metabolism by an integrative "multi-omics" approach: A perspective encompassing genomics, transcriptomics, and proteomics
J Inherit Metab Dis. 2020 Jan. [Epub ahead of print] ***>>PubMed-Link<<***
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Genetic Potassium Channel-Associated Epilepsies: Clinical Review of the Kv Family
Eur J Paediatr Neurol. 2020 Jan [Epub ahead of print] *>>PubMed-Link<<*
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1. Januar 2020 (GAIN)

Distinct molecular response patterns of activating STAT3 mutations associate with penetrance of lymphoproliferation and autoimmunity

Clin Immunol. 2020 Jan [Epub ahead of print 2019 Nov 23] >>PubMed-Link<<

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