

Veröffentlichungen der BMFTR-geförderten Forschungsverbünde für Seltene Erkrankungen, 2024

24. DEZEMBER 2024 (GAIN)

PGM3 insufficiency: a glycosylation disorder causing a notable T cell defect

Front Immunol. 2024 Dec 24 >>[Pubmed-Link](#)<<

Linlin Yang, Barbara Zerbato, Alex Pessina, Luca Brambilla, Virginia Andreani, Stefanie Frey-Jakobs, Manfred Fliegauf, Mohamed-Ridha Barbouche, Qiaoxia Zhang, Ferdinando Chiaradonna, Michele Proietti, Xin Du, Bodo Grimbacher

24. DEZEMBER 2024 (STOP-FSGS)

Ecologically sustainable benchmarking of AI models for histopathology

NPJ Digit Med. 2024 Dec 24. >>[Pubmed-Link](#)<<

Yu-Chia Lan, Martin Strauch, Pourya Pilva, Nikolas E J Schmitz, Alireza Vafaei Sadr, Leon Niggemeier, Huong Quynh Nguyen, David L Hölscher, Tri Q Nguyen, Jesper Kers, Roman D Bülow, Peter Boor

21. DEZEMBER 2024 (NEOCYST)

Pathogenic KIAA0586/TALPID3 variants are associated with defects in primary and motile cilia

iScience. 2024 Dec 21. eCollection 2025 Feb 21 >>[Pubmed-Link](#)<< Jacqueline E Taudien, Diana Bracht, Heike Olbrich, Sebastian Swirski, Fulvio D'Abrusco, Bert Van der Zwaag, Maike Möller, Thomas Lücke, Norbert Teig, Ulrika Lindberg, Kai Wohlgemuth, Julia Wallmeier, Anja Blaque, Christos Gatsogiannis, Sebastian George, Christoph Jüschke, Marta Owczarek-Lipska, Dorothee Veer, Hester Y Kroes, Enza Maria Valente, G Christoph Korenke, Heymut Omran 2, John Neidhardt

20. DEZEMBER 2024 (GAIN)

RNA-binding proteins hnRNPM and ELAVL1 promote type-I interferon induction downstream of the nucleic acid sensors cGAS and RIG-I

EMBO J. 2024 Dec 20. >>[Pubmed-Link](#)<<

Alexander Kirchhoff, Anna-Maria Herzner, Christian Urban, Antonio Piras, Robert Düster, Julia Mahlberg, Agathe Grünewald, Thais M Schlee-Guimarães, Katrin Ciupka, Petro Leka, Robert J Bootz, Christina Wallerath, Charlotte Hunkler, Ann Kristin de Regt, Beate M Kümmerer, Maria Hønholt Christensen, Florian I Schmidt, Min Ae Lee-Kirsch, Claudia Günther, Hiroki Kato, Eva Bartok, Gunther Hartmann, Matthias Geyer, Andreas Pichlmair, Martin Schlee

19. DEZEMBER 2024 (MITONET)

Five-Year Outcomes of Lenadogene Nolpharvovec Gene Therapy in Leber Hereditary Optic Neuropathy

JAMA Ophthalmol. 2024 Dec 19. >>[Pubmed-Link](#)<<

Patrick Yu-Wai-Man, Nancy J Newman, Valérie Biousse, Valerio Carelli, Mark L Moster, Catherine Vignal-Clermont, Thomas Klopstock, Alfredo A Sadun, Robert C Sergott, Rabih Hage, Simona Degli Esposti, Chiara La Morgia, Claudia Priglinger, Rustum Karanja, Magali Tael, José-Alain Sahel; LHON Study Group

18. DEZEMBER 2024 (STOP-FSGS)

A dataset profiling the multiomic landscape of the prefrontal cortex in amyotrophic lateral sclerosis

Gigascience. 2024 Dec 18. >>[Pubmed-Link](#)<<

Fabian Hausmann, Lucas Caldi Gomes, Sonja Hänzelmann, Robin Khatri, Sergio Oller, Marie Gebelin, Mojan Parvaz, Laura Tzeplaeff, Laura Pasetto, Qihui Zhou, Pavol Zelina, Dieter Edbauer, R Jeroen Pasterkamp, Hubert Rehauer, Ralph Schlapbach, Christine Carapito, Valentina Bonetto, Stefan Bonn, Paul Lingor

13. DEZEMBER 2024 (CONNECT-GENERATE)

Frequency, characteristics, and immunological accompaniments of ataxia in anti-NMDAR antibody-associated encephalitis

Front Immunol. 2024 Dec 13. >>[Pubmed-Link](#)<<

Sarah Jesse, Marie Riemann, Hauke Schneider, Marius Ringelstein, Nico Melzer, Niklas Vogel, Lena Kristina Pfeffer, Manuel A Friese, Kurt-Wolfram Sühs, Dominica Hudasch, Philipp Schwenkenbecher, Albrecht Günther, Christian Geis, Jonathan Wickel, Martin Lesser, Annika Kather, Frank Leypoldt, Justina Dargvainiene, Robert Markewitz, Klaus-Peter Wandinger, Franziska S Thaler, Joseph Kuchling, Katharina Wurdack, Lidia Sabater, Carsten Finke, Jan Lewerenz

10. DEZEMBER 2024 (TREAT-ION)

The fruit fly *Drosophila melanogaster* as a screening model for antiseizure medications

Front Pharmacol. 2024 Dec 10. >>[Pubmed-Link](#)<<

Florian P Fischer, Robin A Karge, Henner Koch, Aaron Voigt, Yvonne G Weber, Stefan Wolking

9. DEZEMBER 2024 (STOP-FSGS)

Surgical pathology and sustainable development: international landscape and prospects

J Clin Pathol. 2024 Dec 9. >>[Pubmed-Link](#)<<

Rémi Vergara, Ivan Théate, Peter Boor, Ilyssa O Gordon, Jonathan West, Selma Abdelmoula, Cyprien Tilmant, Roque Gabriel Wiseman Pinto, Lucie Gaillot-Durand, Sheri Scott, Alexis Trecourt, Anne Rullier

9. DEZEMBER 2024 (STOP-FSGS)

ATP-Gated P2X7-Ion Channel on Kidney-Resident Natural Killer T Cells and Memory T Cells in Intrarenal Inflammation

J Am Soc Nephrol. 2024 Dec 9. >>[Pubmed-Link](#)<<

Marten Junge, Nastassia Liaukouskaya, Nicole Schwarz, Carolina Pinto-Espinoza, Alessa Z Schaffrath, Björn Rissiek, Christian F Krebs, Guido Rattay, Hans-Willi Mittrücker, Nicola M Tomas, Annette Nicke, Friedrich Haag, Tobias B Huber, Catherine Meyer-Schwesinger, Friedrich Koch-Nolte, Nicola Wanner

3. DEZEMBER 2024 (ADDRESS)

Re-envisioning genetic predisposition to childhood and adolescent cancers

Nat Rev Cancer. 2024 Dec 3. >>[Pubmed-Link](#)<<

Christian P Kratz

1. DEZEMBER 2024 (ADDRESS)

ERN GENTURIS guidelines on constitutional mismatch repair deficiency diagnosis, genetic counselling, surveillance, quality of life, and clinical management

Eur J Hum Genet. 2024 Dec. >>[Pubmed-Link](#)<<

Chrystelle Colas, Léa Guerrini-Rousseau, Manon Suerink, Richard Gallon, Christian P Kratz, Éloïse Ayuso; ERN GENTURIS CMMRD Guideline Group; Laurence Brugières, Katharina Wimmer

1. DEZEMBER 2024 (MYPRED)

Expanding the genetic landscape of congenital neutropenia: CXCR2 mutations in three families revealed through whole exome sequencing

Haematologica. 2024 Dec 1. >>[Pubmed-Link](#)<<

Maksim Klimiankou, Ivan Tesakov, Grigorios Tsaknakis, Erasmia Boutakoglou, Irene Mavroudi, Malte Ritter, Marc Sturm, Julia Skokowa, Helen A Papadaki

1. DEZEMBER 2024 (MITONET)

Missense variants in the TRPMr7 α -kinase domain are associated with recurrent pediatric acute liver failure

Hepatol Commun. 2024 Nov 29. eCollection 2024 Dec 1. >>[Pubmed-Link](#)<<

Lea D Schlieben, Melanie T Achleitner, Billy Bourke, Max Diesner, René G Feichtinger, Alexander Fichtner, Christa Flechtenmacher, Nedim Hadzic, Robert Hegarty, Andreas Heilos, Andreas Janecke, Vassiliki Konstantopoulou, Dominic Lenz, Johannes A Mayr, Thomas Müller, Holger Prokisch, Georg F Vogel

28. NOVEMBER 2024 (GAIN)

COVID-19 progression and convalescence in common variable immunodeficiency patients show dysregulated adaptive immune responses and persistent type I interferon and inflammasome activation

Nat Commun. 2024 Nov 28. >>[Pubmed-Link](#)<<

Javier Rodríguez-Ubreva, Josep Calafell-Segura, Celia L Calvillo, Baerbel Keller, Laura Ciudad, Louis-François Handfield, Carlos de la Calle-Fabregat, Gerard Godoy-Tena, Eduardo Andrés-León, Regina Hoo, Tarryn Porter, Elena Prigmore, Maike Hofmann, Annegrit Decker, Javier Martín, Roser Vento-Tormo, Klaus Warnatz, Esteban Ballestar

25. NOVEMBER 2024 (CONNECT-GENERATE)

Current and Future Roles of Chimeric Antigen Receptor T-Cell Therapy in Neurology: A Review

JAMA Neurol. 2024 Nov 25. >>[Pubmed-Link](#)<<

Fatme Seval Ismail, Marco Gallus, Sven G Meuth, Hideho Okada, Hans-Peter Hartung, Nico Melzer

23. NOVEMBER 2024 (ADDRESS)

Multi-omic and single-cell profiling of chromothriptic medulloblastoma reveals genomic and transcriptomic consequences of genome instability

Nat Commun. 2024 Nov 23. >>[Pubmed-Link](#)<<

Petr Smirnov, Moritz J Przybilla, Milena Simovic-Lorenz, R Gonzalo Parra, Hana Susak, Manasi Ratnaparkhe, John KI Wong 6, Verena Körber, Jan-Philipp Mallm, George Philippos, Martin Sill, Thorsten Kolb, Rithu Kumar, Nicola Casiraghi, Konstantin Okonechnikov, David R Ghasemi, Kendra Korinna Maaß, Kristian W Pajtler, Anna Jauch, Andrey Korshunov, Thomas Höfer, Marc Zapatka, Stefan M Pfister, Wolfgang Huber, Oliver Stegle, Aurélie Ernst

21. NOVEMBER 2024 (MITONET)

Genotype-specific effects of elamipretide in patients with primary mitochondrial myopathy: a post hoc analysis of the MMPOWER-3 trial

Orphanet J Rare Dis. 2024 Nov 21. >>[Pubmed-Link](#)<<

Amel Karaa, Enrico Bertini, Valerio Carelli, Bruce Cohen, Gregory M Ennes, Marni J Falk, Amy Goldstein, Gráinne Gorman, Richard Haas, Michio Hirano, Thomas Klopstock, Mary Kay Koenig, Cornelia Kornblum, Costanza Lamperti, Anna Lehman, Nicola Longo, Maria Judit Molnar, Sumit Parikh, Han Phan, Robert D S Pitceathly, Russekk Saneto, Fernando Scaglia, Serenella Servidei, Mark Tarnopolsky, Antonio Toscano, Johan L K Van Hove, John Vissing, Jerry Vockley, Jeffrey S Finman, Anthony Abbruscato, David A Brown, Alana Sullivan, James A Shiffer, Michelangelo Mancuso; MMPOWER-3 Trial Investigators

15. NOVEMBER 2024 (ADDRESS)

Update on Recommendations for Cancer Screening and Surveillance in Children with Genomic Instability Disorders

Clin Cancer Res. 2024 Nov 15. >>[Pubmed-Link](#)<<

Yoshiko Nakano, Roland P Kuiper, Kim E Nichols, Christopher C Porter, Harry Lesmana, Julia Meade, Christian P Kratz, Lucy A Godley, Luke D Maese, Maria Isabel Achatz, Payal P Khincha, Sharon A Savage, Andrea S Doria, Mary-Louise C Greer, Vivian Y Chang, Lisa L Wang, Sharon E Plon, Michael F Walsh

13. NOVEMBER 2024 (TREAT-ION)

Ligand distances as key predictors of pathogenicity and function in NMDA receptors

Hum Mol Genet. 2024 Nov 13. >>[Pubmed-Link](#)<<

Ludovica Montanucci, Tobias Brünger, Nisha Bhattarai, Christian M Boßelmann, Sukhan Kim, James P Allen, Jing Zhang, Chiara Klöckner, Ilona Krey, Piero Fariselli, Patrick May, Johannes R Lemke, Scott J Myers, Hongjie Yuan, Stephen F Traynelis, Dennis Lal

12. NOVEMBER 2024 (NEOCYST)

Ultrasound evaluation of kidney and liver involvement in Bardet-Biedl syndrome

Orphanet J Rare Dis. 2024 Nov 12. >>[Pubmed-Link](#)<<

Metin Cetiner, Ilja Finkelberg, Felix Schiepek, Lars Pape, Raphael Hirtz, Anja K Büscher

7. NOVEMBER 2024 (STOP-FSGS)

The role of the tricellular junction protein ILDR2 in glomerulopathies: Expression patterns and functional insights

iScience. 2024 Nov 7. >>[Pubmed-Link](#)<<

Florian Siegerist, Felix Kliewe, Elke Hammer, Paul Schakau, Joanne Ern Chi Soh, Claudia Weber, Maja Lindenmeyer, Simone Reichelt-Wurm, Vedran Drenic, Christos Chatziantoniou, Christos E Chadjichristos, Yiyang Zhang, Stefan Simm, Miriam C Banas, Matthias Nauck, Uwe Völker, Nicole Endlich

6. NOVEMBER 2024 (MITONET)

Phase 2b program with sonlicromanol in patients with mitochondrial disease due to m.3243A>G mutation

Brain. 2024 Nov 6. >>[Pubmed-Link](#)<<

Jan Smeitink, Just van Es, Brigitte Bosman, Mirian C H Janssen, Thomas Klopstock, Grainne Gorman, John Vissing, Gerrit Ruiterkamp, Chris J Edgar, Evertine J Abbink, Rob van Maanen, Oksana Pogoryelova, Claudia Stendel, Almut Bischoff, Ivan Karin, Mahtab Munshi, Anne Kümmel, Lydia Burgert, Christianne Verhaak, Herma Renkema

4. NOVEMBER 2024 (CONNECT-GENERATE)

Flow cytometry identifies changes in peripheral and intrathecal lymphocyte patterns in CNS autoimmune disorders and primary CNS malignancies

J Neuroinflammation. 2024 Nov 4. >>[Pubmed-Link](#)<<

Saskia Räuber, Andreas Schulte-Mecklenbeck, Alice Willison, Ramona Hagler, Marius Jonas, Duygu Pul, Lars Masanneck, Christina B Schroeter, Kristin S Golombeck, Stefanie Lichtenberg, Christine Strippel, Marco Gallus, Andre Dik, Ruth Kerkhoff, Sumanta Barman, Katharina J Weber, Stjepana Kovac, Melanie Korsen, Marc Pawlitzki, Norbert Goebels, Tobias Ruck, Catharina C Gross, Werner Paulus, Guido Reifenberger, Michael Hanke, Oliver Grauer, Marion Rapp, Michael Sabel, Heinz Wiendl, Sven G Meuth, Nico Melzer

2. NOVEMBER 2024 (MYPRED)

Inducible pluripotent stem cell models to study bone marrow failure and MDS predisposition syndromes

Exp Hematol. 2024 Nov 2. >>[Pubmed-Link](#)<<

Sushree S Sahoo, Majd Khiami, Marcin W Wlodarski

1. NOVEMBER 2024 (STOP-FSGS)

Optimized protocol for the multiomics processing of cryopreserved human kidney tissue

Am J Physiol Renal Physiol. 2024 Nov 1. >>[Pubmed-Link](#)<<

Sydney E Gies, Sonja Hänzelmann, Dominik Kylies, Moritz Lassé, Simon Lagies, Fabian Hausmann, Robin Khatri, Nikolay Zolotarev, Manuela Poets, Tianran Zhang, Fatih Demir, Anja M Billing, Josephine Quaas, Elisabeth Meister, Jonas Engesser, Anne K Mühlig, Shun Lu, Shuya Liu, Silvia Chilla, Ilka Edenhofer, Jan Czogalla, Fabian Braun, Bernd Kammerer, Victor G Puelles, Stefan Bonn, Markus M Rinschen, Maja Lindenmeyer, Tobias B Huber

1. NOVEMBER 2024 (STOP-FSGS)

Advances in Molecular Imaging of Kidney Diseases

Nephron. 2024 Nov 1. >>[Pubmed-Link](#)<<

Barbara Mara Klinkhammer, Ilknur Ay, Peter Caravan, Anna Caroli, Peter Boor

1. NOVEMBER 2024 (HICHOL)

Plasma immune signatures can predict rejection-free survival in the first year after pediatric liver transplantation

J Hepatol. 2024 Nov. >>[Pubmed-Link](#)<<

Evgeny Chichelnitskiy, Imeke Goldschmidt, Louisa Ruhl, Nicole Rübsamen, Veronika K Jaeger, Andre Karch, Kerstin Beushausen, Jana Keil, Juliane K Götz, Lorenzo D'Antiga, Dominique Debray, Loreto Hierro, Deirdre Kelly, Valerie McLin, Joanna Pawlowska, Rafael T Mikolajczyk, Michela Bravi, Maja Klaudel-Dreszler, Zeynep Demir, Carla Lloyd, Simona Korff, Ulrich Baumann, Christine S Falk

1. NOVEMBER 2024 (STOP-FSGS)

Inhibition of complement factor C5a or C5aR for cholesterol crystal embolism-related vascular thrombosis with microvascular injury and its consequences

Kidney Int. 2024 Nov. >>[Pubmed-Link](#)<<

Danyang Zhao, Chao Han, Elmina Mammadova-Bach, Kanako Watanabe-Kusunoki, Tamisa Seeko Bandeira Honda, Yihong Li, Chenyu Li, Qiubo Li, Hao Long, Lyuben Lyubenov, Chongxu Shi, Donato Santovito, Christian Weber, Peter Boor, Patrick Droste, Samir Parikh, John Shapiro, Letizia De Chiara, Giulia Carangelo, Paola Romagnani, Sven Klussmann, Kai Hoehlig, Axel Vater, Hans-Joachim Anders

1. NOVEMBER 2024 (STOP-FSGS)

Cyclin-dependent kinase 4 drives cystic kidney disease in the absence of mTORC1 signaling activity

Kidney Int. 2024 Nov. >>[Pubmed-Link](#)<<

Florian Grahammer, Bernhard Dumoulin, Ramila E Gulieva, Hui Wu, Yaoxian Xu, Nurgazy Sulaimanov, Frederic Arnold, Lukas Sandner, Tomke Cordts, Abhijeet Todkar, Pierre Moulin, Wilfried Reichardt, Victor G Puelles, Rafael Kramann, Benjamin S Freedman, Hauke Busch, Melanie Boerries, Gerd Walz, Tobias B Huber

1. NOVEMBER 2024 (CONNECT-GENERATE)

Eigenvector Centrality Mapping Reveals Volatility of Functional Brain Dynamics in Anti-NMDA Receptor Encephalitis

Biol Psychiatry Cogn Neurosci Neuroimaging. 2024 Nov. >>[Pubmed-Link](#)<<

Tim J Hartung, Nina von Schwanenflug, Stephan Krohn, Tommy A A Broeders, Harald Prüss, Menno M Schoonheim, Carsten Finke

1. NOVEMBER 2024 (CONNECT-GENERATE)

Strategies to improve autoimmune neurological diseases treatment

Rev Neurol (Paris). 2024 Nov. >>[Pubmed-Link](#)<<

F Leypoldt

1. NOVEMBER 2024 (CONNECT-GENERATE)

MRI findings in autoimmune encephalitis

Rev Neurol (Paris). 2024 Nov. >>[Pubmed-Link](#)<<

T J Hartung, F Bartels, J Kuchling, S Krohn, J Leidel, M Mantwill, K Wurdack, S Yogeshwar, M Scheel, C Finke

24. OKTOBER 2024 (HICHOL)

ECM1 attenuates hepatic fibrosis by interfering with mediators of latent TGF- β 1 activation

Gut. 2024 Oct 24. >>[Pubmed-Link](#)<<

Frederik Link, Yujia Li, Jieling Zhao, Stefan Munker, Weiguo Fan, Zeribe C Nwosu, Ye Yao, Shanshan Wang, Chenjun Huang, Roman Liebe, Seddik Hammad, Hui Liu, Chen Shao, Chunfang Gao, Bing Sun, Natalie J Török, Huiguo Ding, Matthias Pa Ebert, Honglei Weng, Peter Ten Dijke, Dirk Drasdo, Steven Dooley, Sai Wang

22. OKTOBER 2024 (TREATHSP.NET)

Loss of CLN3 in microglia leads to impaired lipid metabolism and myelin turnover

Commun Biol. 2024 Oct 22. >>[Pubmed-Link](#)<<

Seda Yasa, Elisabeth S Butz, Alessio Colombo, Uma Chandrachud, Luca Montore, Sarah Tschirner, Matthias Prestel, Steven D Sheridan, Stephan A Müller, Janos Groh, Stefan F Lichtenthaler, Sabina Tahirovic, Susan L Cotman

10. OKTOBER 2024 (ADDRESS)

Clustering of TP53 variants into functional classes correlates with cancer risk and identifies different phenotypes of Li-Fraumeni syndrome

KI Rep. 2025 Jan. Epub 2024 Oct 10. >>[Pubmed-Link](#)<<

Emilie Montellier, Nathanaël Lemonnier, Judith Penkert, Claire Freycon, Sandrine Blanchet, Amina Amadou, Florent Chuffart, Nicholas W Fischer, Maria-Isabel Achatz, Arnold J Levine, Catherine Goudie, David Malkin, Gaëlle Bougeard, Christian P Kratz, Pierre Hainaut

1. OKTOBER 2024 (STOP-FSGS)

Pathophysiologic Mapping of Chronic Liver Diseases With Longitudinal Multiparametric MRI in Animal Models

Invest Radiol. 2024 Oct 1. >>[Pubmed-Link](#)<<

Ferhan Baskaya, Teresa Lemainque, Barbara Klinkhammer, Susanne Koletnik, Saskia von Stillfried, Steven R Talbot, Peter Boor, Volkmar Schulz, Wiltrud Lederle, Fabian Kiessling

1. OKTOBER 2024 (GAIN)

Phenotypic and pathomechanistic overlap between tapasin and TAP deficiencies

J Allergy Clin Immunol. 2024 Oct. >>[Pubmed-Link](#)<<

Abdulwahab Elsayed, Sandra von Hardenberg, Faranaz Atschekzei, Theresa Graalmann, Christine Jänke, Torsten Witte, Felix C Ringshausen, Georgios Sogkasg

1. OKTOBER 2024 (MYPRED)

Comparison of Gene-Editing Approaches for Severe Congenital Neutropenia-Causing Mutations in the ELANE Gene

CRISPR J. 2024 Oct. >>[Pubmed-Link](#)<<

Malte Ulrich Ritter, Masoud Nasri, Benjamin Dannenmann, Perihan Mir, Benjamin Secker, Diana Amend, Maksim Klimiankou, Karl Welte, Julia Skokowa

1. OKTOBER 2024 (CONNECT-GENERATE)

Antibodies in neurological diseases: Established, emerging, explorative

Immunol Rev. 2024 Oct 1. >>[Pubmed-Link](#)<<

Lucie Y Li, Amelya Keles, Marie A Homeyer, Harald Prüss

25. SEPTEMBER 2024 (STOP-FSGS)

Targeting Kidney Fibrosis with Polymeric Nanocarriers

Chem Mater. 2024 Sept 25. >>[ASC Publications-Link](#)<<

Vedangi Kulkarni, Tim Caspers, Diana Moeckel, Susanne Koletnik, Elena Rama, Paraskevi Sotiropoulou, Daniel R. Dietrich, Robert Pola, Lenka Kotrchová, Lenka Kotrchová, Tomáš Etrych, Yvonne H. W. Derks, Peter Boor, Fabian Kiessling, Felix M. Mottaghy, Barbara M. Klinkhammer, Twan Lammers

18. SEPTEMBER 2024 (HICHOL)

Gut microbiota-derived butyrate selectively interferes with growth of carbapenem-resistant Escherichia coli based on their resistance mechanism

Gut Microbes. 2024 Jan-Dec (Sept. 18). >>[Pubmed-Link](#)<<

Eva Happ, Kora Schulze, Zinia Afrin, Sabrina Woltemate, Pia Görner, Stefan Ziesing, Dirk Schlüter, Robert Geffers, Volker Winstel, Marius Vital

17. SEPTEMBER 2024 (STOP-FSGS)

ADAMTS12 promotes fibrosis by restructuring extracellular matrix to enable activation of injury-responsive fibroblasts

J Clin Invest. 2024 Sep 17. >>[Pubmed-Link](#)<<

Konrad Hoeft, Lars Koch, Susanne Ziegler, Ling Zhang, Steffen Luetke, Maria C Tanzer, Debashish Mohanta, David Schumacher, Felix Schreiebing, Qingqing Long, Hyojin Kim, Barbara M Klinkhammer, Carla Schikarski, Sidrah Maryam, Mathijs Baens, Juliane Hermann, Sarah Krieg, Fabian Peisker, Laura De Laporte, Gideon JI Schaefer, Sylvia Menzel, Joachim Jankowski, Benjamin D Humphreys, Adam Wahida, Rebekka K Schneider, Matthias Versele, Peter Boor, Matthias Mann, Gerhard Sengle, Sikander Hayat, Rafael Kramann

12. SEPTEMBER 2024 (GAIN)

Monogenic lupus – from gene to targeted therapy

Mol Cell Pediatr. 2024 Sep 12. >>[Pubmed-Link](#)<<

Katharina Menzel, Kateryna Novotna, Nivya Jeyakumar, Christine Wolf, Min Ae Lee-Kirsch

8. SEPTEMBER 2024 (STOP-FSGS)

Improved health by combining dietary restriction and promoting muscle growth in DNA repair-deficient progeroid mice

J Cachexia Sarcopenia Muscle. 2024 Sep 8. >>[Pubmed-Link](#)<<

Wilbert P Vermeij, Khalid Alyodawi, Ivar van Galen, Jennie L von der Heide, María B Birkisdóttir, Lisanne J Van't Sant, Rutger A Ozinga, Daphne S J Komninos, Kimberly Smit, Yvonne M A Rijksen, Renata M C Brandt, Sander Barnhoorn, Dick Jaarsma, Sathivel Vaiyapuri, Olli Ritvos, Tobias B Huber, Oliver Kretz, Ketan Patel

3. SEPTEMBER 2024 (CONNECT-GENERATE)

Structural and functional mechanisms of anti-NMDAR autoimmune encephalitis

Nat Struct Mol Biol. 2024 Sep 3. >>[Pubmed-Link](#)<<

Kevin Michalski, Taha Abdulla, Sam Kleeman, Lars Schmidl, Ricardo Gómez, Noriko Simorowski, Francesca Vallese, Harald Prüss, Manfred Heckmann, Christian Geis, Hiro Furukawa

2. SEPTEMBER 2024 (CONNECT-GENERATE)

Autoantigen-specific CD4+ T cells acquire an exhausted phenotype and persist in human antigen-specific autoimmune diseases

Immunity. 2024 Sept 2. >>[Pubmed-Link](#)<<

Carina Saggau, Petra Bacher, Daniela Esser, Mahdi Rasa, Silja Meise, Nicola Mohr, Nora Kohlstedt, Andreas Hutloff, Sarah-Sophie Schacht, Justina Dargvainiene, Gabriela Rios Martini, Klarissa H. Stürner, Ina Schröder, Robert Markewitz, Johannes Hartl, Maria Hastermann, Ankelien Duchow, Patrick Schindler, Mareike Becker, Carolin Bautista, Judith Gottfreund, Jörn Walter, Julia K. Polansky, Mingxing Yang, Reza Naghavian, Mareike Wendorff, Ev-Marie Schuster, Andreas Dahl, Andreas Petzold, Susanne Reinhardt, Andre Franke, Marek Wieczorek, Lea Henschel, Daniel Berger, Guido Heine, Maïke Holtsche, Vivien Häußler, Christian Peters, Enno Schmidt, Simon Fillatreau, Dirk H. Busch, Klaus-Peter Wandinger, Kilian Schober, Roland Martin, Friedemann Paul, Frank Leypoldt, Alexander Scheffold

1. SEPTEMBER 2024 (GAIN)

Novel hypermorphic variants in IRF2BP2 identified in patients with common variable immunodeficiency and autoimmunity

Clin Immunol. 2024 Sep. >>[Pubmed-Link](#)<<

Manfred Anim, Georgios Sogkas, Nadezhda Camacho-Ordonez, Gunnar Schmidt, Abdulwahab Elsayed, Michele Proietti, Torsten Witte, Bodo Grimbacher, Faranaz Atschekzei

1. SEPTEMBER 2024 (CONNECT-GENERATE)

LGI1 Autoantibodies Enhance Synaptic Transmission by Presynaptic Kv1 Loss and Increased Action Potential Broadening

Neurol Neuroimmunol Neuroinflamm. 2024 Sep. >>[Pubmed-Link](#)<<

Andreas Ritzau-Jost, Felix Gsell, Josefine Sell, Stefan Sachs, Jacqueline Montanaro, Toni Kirmann, Sebastian Maaß, Sarosh R Irani, Christian Werner, Christian Geis, Markus Sauer, Ryuichi Shigemoto, Stefan Hallermann

1. SEPTEMBER 2024 (STOP-FSGS)

Super-resolved highly multiplexed immunofluorescence imaging for precise protein localization and podocyte ultrastructure

J Cell Mol Med. 2024 Sep. >>[Pubmed-Link](#)<<

Florian Siegerist, Svenja Kitzel, Nihal Telli, Juan Saydou Dikou, Vedran Drenić, Christos E ChadjiChristos, Christos Chatziantoniou, Nicole Endlich

30. AUGUST 2024 (CONNECT-GENERATE)

Progressive Encephalomyelitis With Rigidity and Myoclonus With Glycine Receptor and GAD65 Antibodies: Case Report and Potential Mechanisms

Neurol Neuroimmunol Neuroinflamm. 2024 Nov. Epub 2024 Aug 30. >>[Pubmed-Link](#)<<

Michael Winklehner, Jonathan Wickel, Ellen Gelpi, Dirk Brämer, Vera Rauschenberger, Albrecht Günther, Jan Bauer, Anika Simonovska Serra, Philipp Jauk, Carmen Villmann, Romana Höftberger, Christian Geis

23. AUGUST 2024 (GAIN)

JAK inhibitors to treat STAT3 gain-of-function: a single-center report and literature review

Front Immunol. 2024 Aug 23. >>[Pubmed-Link](#)<<

Faranaz Atschekzei, Stephan Traidl, Julia Carlens, Katharina Schütz, Sandra von Hardenberg, Abdulwahab Elsayed, Diana Ernst, Linus Risser, Thea Thiele, Theresa Graalman, Juliana Raab, Ulrich Baumann, Torsten Witte, Georgios Sogkas

17. AUGUST 2024 (STOP-FSGS)

3D Humanized Bioprinted Tubulointerstitium Model to Emulate Renal Fibrosis In Vitro

Adv Healthc Mater. 2024 Aug 17. >>[Pubmed-Link](#)<<

Gabriele Addario, Julia Fernández-Pérez, Chiara Formica, Konstantinos Karyniotakis, Lea Herkens, Sonja Djudjaj, Peter Boor, Lorenzo Moroni, Carlos Mota

15. AUGUST 2024 (STOP-FSGS)

Adeno-associated virus-based gene therapy treats inflammatory kidney disease in mice

J Clin Invest. 2024 Aug 15. >>[Pubmed-Link](#)<<

Guochao Wu, Shuya Liu, Julia Hagenstein, Malik Alawi, Felicitas E Hengel, Melanie Schaper, Nuray Akyüz, Zhouning Liao, Nicola Wanner, Nicola M Tomas, Antonio Virgilio Failla, Judith Dierlamm, Jakob Körbelin, Shun Lu, Tobias B Huber

14. AUGUST 2024 (TREATHSP.NET)

Alpha-synuclein fine-tunes neuronal response to pro-inflammatory cytokines

Brain Behav Immun. 2024 Aug 14. >>[Pubmed-Link](#)<<

Veronika Sigutova, Wei Xiang, Martin Regensburger, Beate Winner, Iryna Prots

13. AUGUST 2024 (GENERARE)

Impaired Intracellular Calcium Buffering Contributes to the Arrhythmogenic Substrate in Atrial Myocytes From Patients With Atrial Fibrillation

Circulation. 2024 Aug 13. >>[Pubmed-Link](#)<<

Funsho E Fakuade, Dominik Hubricht, Vanessa Möller, Izzatullo Sobitov, Aiste Liutkute, Yannic Döring, Fitzwilliam Seibert, Marcus Gerloff, Julius Ryan D Pronto, Fereshteh Haghighi, Sören Brandenburg, Khaled Alhussini, Nadezda Ignatyeva, Yara Bonhoff, Stefanie Kestel, Aschraf El-Essawi, Ahmad Fawad Jebran, Marius Großmann, Bernhard C Danner, Hassina Baraki, Constanze Schmidt, Samuel Sossalla, Ingo Kutschka, Constanze Bening, Christoph Maack, Wolfgang A Linke, Jordi Heijman, Stephan E Lehnart, George Kensah, Antje Ebert, Fleur E Mason, Niels Voigt

13. AUGUST 2024 (CONNECT-GENERATE)

Human cerebrospinal fluid monoclonal CASPR2 autoantibodies induce changes in electrophysiology, functional MRI, and behavior in rodent models

Brain Behav Immun. 2024 Aug 13. >>[Pubmed-Link](#)<<

Scott van Hoof, Jakob Kreye, César Cordero-Gómez, Julius Hoffmann, S Momen Reincke, Elisa Sánchez-Sendin, Sophie L Duong, Manoj Upadhyay, Divya Dhangar, Paulina Michór, Gavin L Woodhall, Maraike Küpper, Andreas Oder, Joseph Kuchling, Stefan Paul Koch, Susanne Mueller, Philipp Boehm-Sturm, Jens Peter von Kries, Carsten Finke, Timo Kirschstein, Sukhvir K Wright, Harald Prüss

2. AUGUST 2024 (CONNECT-GENERATE)

High serum prevalence of autoreactive IgG antibodies against peripheral nerve structures in patients with neurological post-COVID-19 vaccination syndrome

Front Immunol. 2024 Aug 2. >>[Pubmed-Link](#)<<

Friederike A Arlt, Ameli Breuer, Elli Trampenau, Fabian Boesl, Marieluise Kirchner, Philipp Mertins, Elisa Sánchez-Sendín, Mahoor Nasouti, Marie Mayrhofer, Martin Blüthner, Matthias Endres, Harald Prüss, Christiana Franke

1. AUGUST 2024 (MYPRED)

SH2B3 alterations in a novel genetic condition, juvenile myelomonocytic leukemia, and myeloproliferative neoplasia

Haematologica. 2024 Aug 1. >>[Pubmed-Link](#)<< (Editorial)

Charlotte M Niemeyer, Miriam Erlacher

1. AUGUST 2024 (TREATHSP.NET)

Mobile digital gait analysis captures effects of botulinum toxin in hereditary spastic paraplegia

Eur J Neurol. 2024 Aug. >>[Pubmed-Link](#)<<

Alzhrara A Ibrahim, Malte Ollenschläger, Stephan Klebe, Rebecca Schüle, Nicole Jeschonneck, Melanie Kellner, Evelyn Loris, Teresa Greinwalder, Bjoern M Eskofier, Jürgen Winkler, Heiko Gaßner, Martin Regensburger

1. AUGUST 2024 (GAIN)

OTULIN-related conditions: Report of a new case and review of the literature using GenIA

Clin Immunol. 2024 Aug. >>[Pubmed-Link](#)<<

Andrés Caballero-Oteyza, Laura Crisponi, Xiao P Peng, Hongying Wang, Pavla Mrovecova, Stefania Olla, Chiara Siguri, Farida Marnissi, Zineb Jouhadi, Ivona Aksentijevich, Bodo Grimbacher, Michele Proietti

1. AUGUST 2024 (STOP-FSGS)

DNA-binding protein-A: a multitool in tubular epithelial cells

Kidney Int. 2024 Aug. >>[Pubmed-Link](#)<<

Patrick Droste, Peter Boor, Roman D Bülow

31. JULI 2024 (NEOCYST)

Bardet-Biedl syndrome improved diagnosis criteria and management: Inter European Reference Networks consensus statement and recommendations

Eur J Hum Genet. 2024 Jul 31. >>[Pubmed-Link](#)<<

Hélène Dollfus, Marc R Lilien, Pietro Maffei, Alain Verloes, Jean Muller, Giacomo M Bacci, Metin Cetiner, Erica L T van den Akker, Monika Grudzinska Pechhacker, Francesco Testa, Didier Lacombe, Marijn F Stokman, Francesca Simonelli, Aurélie Gouronc, Amélie Gavard, Mieke M van Haelst, Jens Koenig, Sylvie Rossignol, Carsten Bergmann, Miriam Zacchia, Bart P Leroy, Hélène Mosbah, Albertien M Van Eerde, Djalila Mekahli, Aude Servais, Christine Poitou, Diana Valverde

30. JULI 2024 (HICHOL)

Investigating the response of the butyrate production potential to major fibers in dietary intervention studies

NPJ Biofilms Microbiomes. 2024 Jul 30. >>[Pubmed-Link](#)<<

Thao Van-Wehle, Marius Vital

23. JULI 2024 (GAIN)

A novel transmembrane CXCR4 variant that expands the WHIM genotype-phenotype paradigm

Blood Adv. 2024 Jul 23. >>[Pubmed-Link](#)<<

Katarina Zmajkovicova, Sumit Pawar, Svetlana O Sharapova, Christoph B Geier, Ivana Wiest, Chi Nguyen, Halenya Monticelli, Sabine Maier-Munsa, Kelly Chen, John W Sleasman, Svetlana Aleshkevich, Ekaterina Polyakova, Inga Sakovich, Klaus Warnatz, Bodo Grimbacher, Michele Proietti, Neal Sondheimer, Boglarka Ujhazi, Sumai Gordon, Maryssa Ellison, Melis Yilmaz, Jolan E Walter, Adriana Badarau, Arthur G Taveras, Jadee L Neff, Jacob R Bledsoe, Teresa K Tarrant

22. JULI 2024 (STOP-FSGS)

Long-term outcomes of patients with IgA nephropathy in the German CKD cohort

Clin Kidney J. 2024 Jul 22. >>[Pubmed-Link](#)<<

Eleni Stamellou, Jennifer Nadal, Bruce Hendry, Alex Mercer, Claudia Seikrit, Wibke Bechtel-Walz, Matthias Schmid, Marcus J Moeller, Mario Schiffer, Kai-Uwe Eckardt, Rafael Kramann, Jürgen Floege; GCKD study investigators

18. JULI 2024 (ADDRESS)

Clonal relapse dynamics in acute myeloid leukemia following allogeneic hematopoietic cell transplantation

Blood. 2024 Jul 18. >>[Pubmed-Link](#)<<

Clara Philine Wienecke, Bennet Heida, Letizia Venturini, Razif Gabdoulline, Katja Krüger, Katrin Teich, Konstantin Büttner, Martin Wichmann, Wolfram Puppe, Blerina Neziri, Marlene Reuter, Elke Dammann, Michael Stadler, Arnold Ganzer, Lothar Hambach, Felicitas Thol, Michael Heuser

16. JULI 2024 (STOP-FSGS)

Deep learning applications in digital pathology

Nat Rev Nephrol. 2024 Jul 16. >>[Pubmed-Link](#)<<

Peter Boor

10. JULI 2024 (CONNECT-GENERATE)

HLA and KIR genetic association and NK cells in anti-NMDAR encephalitis

Front Immunol. 2024 Jul 10. >>[Pubmed-Link](#)<<

Vicente Peris Sempere, Guo Luo, Sergio Muñoz-Castrillo, Anne-Laurie Pinto, Géraldine Picard, Véronique Rogemond, Maarten J Titulaer, Carsten Finke, Frank Leypoldt, Gregor Kuhlenbäumer; GENERATE study group; Hannah F Jones, Russell C Dale, Sophie Binks, Sarosh R Irani, Anna E Bastiaansen, Juna M de Vries, Marienke A A M de Bruijn, Dave L Roelen, Tae-Joon Kim, Kon Chu, Soon-Tae Lee, Takamichi Kanbayashi, Nicholas R Pollock, Katherine M Kichula, Abigail Mumme-Monheit, Jérôme Honnorat, Paul J Norman, Emmanuel Mignot

5. JULI 2024 (CONNECT-GENERATE)

HLA-DQB1*05 subtypes and not DRB1*10:01 mediates risk in anti-IgLON5 disease

Brain. 2024 Jul 5. >>[Pubmed-Link](#)<<

Selina M Yogeshwar, Sergio Muñoz-Castrillo, Lidia Sabater, Vicente Peris-Sempere, Vamsee Mallajosyula, Guo Luo, Han Yan, Eric Yu, Jing Zhang, Ling Lin, Flavia Fagundes Bueno, Xuhuai Ji, Géraldine Picard, Véronique Rogemond, Anne Laurie Pinto, Anna Heidbreder, Romana Höftberger, Francesc Graus, Josep Dalmau, Joan Santamaria, Alex Iranzo, Bettina Schreiner, Maria Pia Giannoccaro, Rocco Liguori, Takayoshi Shimohata, Akio Kimura, Yoya Ono, Sophie Binks, Sara Mariotto, Alessandro Dinoto, Michael Bonello, Christian J Hartmann, Nicola Tambasco, Pasquale Nigro, Harald Prüss, Andrew McKeon, Mark M Davis, Sarosh R Irani, Jérôme Honnorat, Carles Gaig, Carsten Finke, Emmanuel Mignot

5. JULI 2024 (TREAT-ION)

Persistent sodium currents in neurons: potential mechanisms and pharmacological blockers

Pflugers Arch. 2024 Jul 5. >>[Pubmed-Link](#)<<

Peter Müller, Andreas Draguhn, Alexei V Egorov

3. JULI 2024 (HICHOL)

Prediction of anastomotic insufficiency based on the mucosal microbiome prior to colorectal surgery: a proof-of-principle study

Sci Rep. 2024 Jul 3. >>[Pubmed-Link](#)<<

Konrad Lehr, Undine Gabriele Lange, Noam Mathias Hipler, Ramiro Vilchez-Vargas, Albrecht Hoffmeister, Jürgen Feisthammel, Dorina Buchloh, Denny Schanze, Martin Zenker, Ines Gockel, Alexander Link, Boris Jansen-Winkeln

1. JULI 2024 (TREAT-ION)

Cenobamate: real-world data from a retrospective multicenter study

J Neurol. 2024 Jul 1. >>[Pubmed-Link](#)<<

Stephan Lauxmann, David Heuer, Jan Heckelmann, Florian P Fischer, Melanie Schreiber, Elisabeth Schriewer, Guido Widman, Yvonne Weber, Holger Lerche, Michael Alber, Sigrid Schuh-Hofer, Stefan Wolking

1. JULI 2024 (STOP-FSGS)

Long-term outcomes of adults with FSGS in the German Chronic Kidney Disease cohort

Clin Kidney J. 2024 Apr 27. eCollection 2024 Jul. >>[Pubmed-Link](#)<<

Eleni Stamellou, Jennifer Nadal, Bruce Hendry, Alex Mercer, Wibke Bechtel-Walz, Mario Schiffer, Kai-Uwe Eckardt, Rafael Kramann, Marcus J Moeller, Jürgen Floege; GCKD study investigators

1. JULI 2024 (STOP-FSGS)

Plasmacytoid dendritic cells control homeostasis of megakaryopoiesis

Nature. 2024 Jul. >>[Pubmed-Link](#)<<

Florian Gaertner, Hellen Ishikawa-Ankerhold, Susanne Stutte, Wenwen Fu, Jutta Weitz, Anne Dueck, Bhavishya Nelakuditi, Valeria Fumagalli, Dominic van den Heuvel, Larissa Belz, Gulnoza Sobirova, Zhe Zhang, Anna Titova, Alejandro Martinez Navarro, Kami Pekayvaz, Michael Lorenz, Louisa von Baumgarten, Jan Kranich, Tobias Straub, Bastian Popper, Vanessa Zheden, Walter Anton Kaufmann, Chenglong Guo, Guido Piontek, Saskia von Stillfried, Peter Boor, Marco Colonna, Sebastian Clauß, Christian Schulz, Thomas Bocker, Barbara Walzog, Christoph Scheiermann, William C Aird, Claus Nerlov, Konstantin Stark, Tobias Petzold, Stefan Engelhardt, Michael Sixt, Robert Hauschild, Martina Rudelius, Robert A J Oostendorp, Matteo Iannacone, Matthias Heinig, Steffen Massberg

7. JUNI 2024 (STOP-FSGS)

Multimic ALS signatures highlight subclusters and sex differences suggesting the MAPK pathway as therapeutic target

Nat Commun. 2024 Jun 7. >>[Pubmed-Link](#)<<

Lucas Caldi Gomes, Sonja Hänzelmann, Fabian Hausmann, Robin Khatri, Sergio Oller, Mojan Parvaz, Laura Tzeplaeff, Laura Pasetto, Marie Gebelin, Melanie Ebbing, Constantin Holzapfel, Stefano Fabrizio Columbro, Serena Scozzari, Johanna Knöferle, Isabell Cordts, Antonia F Demleitner, Marcus Deschauer, Claudia Dufke, Marc Sturm, Qihui Zhou, Pavol Zelina, Emma Sudria-Lopez, Tobias B Haack, Sebastian Streb, Magdalena Kuzma-Kozakiewicz, Dieter Edbauer, R Jeroen Pasterkamp, Endre Laczko, Hubert Rehauer, Ralph Schlapbach, Christine Carapito, Valentina Bonetto, Stefan Bonn, Paul Lingor

5. JUNI 2024 (MYPRED)

CRISPR-Cas9n-mediated ELANE promoter editing for gene therapy of severe congenital neutropenia

Mol Ther. 2024 Jun 5. >>[Pubmed-Link](#)<<

Masoud Nasri, Malte U Ritter, Perihan Mir, Benjamin Dannenmann, Masako M Kaufmann, Patricia Arreba-Tutusaus, Yun Xu, Natalia Borbaran-Bravo, Maksim Klimiankou, Claudia Lengerke, Cornelia Zeidler, Toni Cathomen, Karl Welte, Julia Skokowa

4. JUNI 2024 (TREATHSP.NET)

Late-onset Krabbe disease presenting as spastic paraplegia – implications of GCase and CTSB/D

Ann Clin Transl Neurol. 2024 Jun 4. >>[Pubmed-Link](#)<<

Rebecca Mächtel, Jan-Philipp Dobert, Ute Hehr, Alexander Weiss, Matthias Kettwig, Lucia Laugwitz, Samuel Groeschel, Manuel Schmidt, Philipp Arnold, Martin Regensburger, Friederike Zünke

3. JUNI 2024 (ADDRESS)

Update on Cancer Predisposition Syndromes and Surveillance Guidelines for Childhood Brain Tumors

Clin Cancer Res. 2024 Jun 3. >>[Pubmed-Link](#)<<

Jordan R Hansford, Anirban Das, Rose B McGee, Yoshiko Nakano, Jack Brzezinski, Sarah R Scollon, Surya P Rednam, Jaclyn Schiend, Orli Michaeli, Sun Young Kim, Mary-Louise C Greer, Rosanna Weksberg, Douglas R Stewart, William D Foulkes, Uri Tabori, Kristian W Pajtler, Stefan M Pfister, Garrett M Brodeur, Junne Kamihara

1. JUNI 2024 (NEOCYST)

Development of a tool for predicting HNF1B mutations in children and young adults with congenital anomalies of the kidneys and urinary tract

Pediatr Nephrol. 2024 Jun. >>[Pubmed-Link](#)<<

Marcin Kołbuc, Mateusz F Kołek, Rafał Motyka, Beata Bieniaś, Sandra Habbig, Kathrin Burgmaier, Larisa Prikhodina, Svetlana Papizh, Velibor Tasic, Christine Okorn, Maria Szczepańska, Katarzyna Kiliś-Pstrusińska, Anna Wasilewska, Piotr Adamczyk, Marcin Tkaczyk, Małgorzata Pańczyk-Tomaszewska, Monika Miklaszewska, Krzysztof Pawlaczyk, Ewelina Bukowska-Olech, Aleksander Jamsheer, Augustina Jankauskiene, Jens König, Hae Il Cheong, Yo Han Ahn, Sophie Kaspar, Przemysław Sikora, Bodo B Beck, Marcin Zaniew

1. JUNI 2024 (TREAT-ION)

Retigabine and gabapentin restore channel function and neuronal firing in a cellular model of an epilepsy-associated dominant-negative KCNQ5 variant

Neuropharmacology. 2024 Jun 1. >>[Pubmed-Link](#)<<

Johanna Krüger, Holger Lerche

1. JUNI 2024 (MYPRED)

Cytogenetics and genomics in CML and other myeloproliferative neoplasms

Best Practice & Research Clinical Haematology. 2024 June . >>[ScienceDirect-Link](#)<<

Hans H. Kreipe, Brigitte Schlegelberger

27. MAI 2024 (NEOCYST)

Pathogenic PHIP Variants are Variably Associated With CAKUT

Kidney Int Rep. 2024 May 27. eCollection 2024 Aug. >>[Pubmed-Link](#)<<

Jonathan de Fallois, Tobias Sieckmann, Ria Schönauer, Friederike Petzold, Johannes Münch, Melissa Pauly, Georgia Vasileiou, Christin Findeisen, Antje Kampmeier, Alma Kuechler, André Reis, Eva Decker, Carsten Bergmann, Konrad Platzer, Velibor Tasic, Karin Michaela Kirschner, Shirlee Shril, Friedhelm Hildebrandt, Wendy K Chung, Jan Halbritter

25. MAI 2024 (STOP-FSGS)

Autoantibodies Targeting Nephritin in Podocytopathies

N Engl J Med. 2024 May 25. >>[Pubmed-Link](#)<<

Felicitas E Hengel, Silke Dehde, Moritz Lassé et al.

22. MAI 2024 (GAIN)

Variable Syndromic Immunodeficiency in Patients with Biallelic PRIM1 Mutations

J Clin Immunol. 2024 May 22. >>[Pubmed-Link](#)<<

Vasil Toskov, Petra Kaiser-Labusch, Min Ae Lee-Kirsch; PRIM1 study group; Stephan Ehl, Oliver Wegehaupt

22. MAI 2024 (MITONET)

Dystonia and mitochondrial disease: the movement disorder connection revisited in 900 genetically diagnosed patients

J Neurol. 2024 May 22. >>[PubMed-Link](#)<<

Elisabetta Indelicato, Lea D Schlieben, Sarah L Stenton, Sylvia Boesch, Matej Skorvanek, Jan Necpal, Robert Jech, Juliane Winkelmann, Holger Prokisch, Michael Zech

17. MAI 2024 (STOP-FSGS)

A prognostic neural epigenetic signature in high-grade glioma

Nat Med. 2024 May 17. >>[PubMed-Link](#)<<

Richard Drexler, Robin Khatri, Thomas Sauvigny, Malte Mohme, Cecile L Maire, Alice Ryba, Yahya Zghaibeh, Lasse Dührsen, Amanda Salviano-Silva, Katrin Lamszus, Manfred Westphal, Jens Gempt, Annika K Wefers, Julia E Neumann, Helena Bode, Fabian Hausmann, Tobias B Huber, Stefan Bonn,

Kerstin Jütten, Daniel Delev, Katharina J Weber, Patrick N Harter, Julia Onken, Peter Vajkoczy, David Capper, Benedikt Wiestler, Michael Weller, Berend Snijder, Alicia Buck, Tobias Weiss, Pauline C Göller, Felix Sahm, Joelle Aline Menstel, David Niklas Zimmer, Michael B Keough, Lijun Ni, Michelle Monje, Dana Silverbush, Volker Hovestadt, Mario L Suvà, Saritha Krishna, Shawn L Hervey-Jumper, Ulrich Schüller, Dieter H Heiland, Sonja Hänzelmann, Franz L Ricklefs

16. MAI 2024 (CONNECT-GENERATE)

HLA dependency and possible clinical relevance of intrathecally synthesized anti-IgLON5 IgG4 in anti-IgLON5 disease

Front Immunol. 2024 May 16. >>[PubMed-Link](#)<<

Inga Koneczny, Stefan Macher, Markus Hutterer, Thomas Seifert-Held, Evelyn Berger-Sieczkowski, Morten Blaabjerg, Markus Breu, Jens Dreyhaupt, Livia Almeida Dutra, Marcus Erdler, Ingrid Fae, Gottfried Fischer, Florian Frommlet, Anna Heidbreder, Birgit Högl, Veronika Klose, Sigrid Klotz, Herburg Liendl, Mette S Nissen, Jasmin Rahimi, Raphael Reinecke, Gerda Ricken, Ambra Stefani, Marie Süße, Helio A G Teive, Serge Weis, Thomas Berger, Lidia Sabater, Carles Gaig, Jan Lewerenz, Romana Höftberger

16. MAI 2024 (NEOCYST)

Renal and Extrarenal Phenotypes in Patients With HNF1B Variants and Chromosome 17q12 Microdeletions

Kidney Int Rep. 2024 May 16. eCollection 2024 Aug. >>[Pubmed-Link](#)<<

Bénédicte Buffin-Meyer, Juliette Richard, Vincent Guignon, Stefanie Weber, Jens König, Laurence Heidet, Nabila Moussaoui, Jeanne-Pierrette Vu, Stanislas Faguer, Audrey Casemayou, Richa Prakash, Véronique Baudouin, Julien Hogan, Demi Alexandrou, Detlef Bockenhauer, Justine Bacchetta, Bruno Ranchin, Stepanka Pruhova, Jakub Zieg, Annie Lahoche, Christine Okorn, Violetta Antal-Kónya, Denis Morin, Francesca Becherucci, Sandra Habbig, Max C Liebau, Mathilde Mauras, Tom Nijenhuis, Brigitte Llanas, Djalila Mekahli, Julia Thumfart, Burkhard Tönshoff, Laura Massella, Philippe Eckart, Sylvie Cloarec, Alejandro Cruz, Ludwig Patzer, Gwenaëlle Roussey, Isabelle Vrillon, Olivier Dunand, Lucie Bessenay, Francesca Taroni, Marcin Zaniew, Ferialle Louillet, Carsten Bergmann, Franz Schaefer, Albertien M van Eerde, Joost P Schanstra, Stéphane Decramer; HNF1B variant study group

1. MAI 2024 (STOP-FSGS)

The role of desmoglein-2 in kidney disease

Kidney Int. 2024 May. >>[Pubmed-Link](#)<<

Tong Xu, Lea Herkens, Ting Jia, Barbara M Klinkhammer, Sebastian Kant, Claudia A Krusche, Eva M Buhl, Sikander Hayat, Jürgen Floege, Pavel Strnad, Rafael Kramann, Sonja Djudjaj, Peter Boor

1. MAI 2024 (ADDRESS)

Adult-Onset Cancer Predisposition Syndromes in Children and Adolescents-To Test or not to Test?

Clin Cancer Res. 2024 May 1. >>[Pubmed-Link](#)<<

Christian P Kratz, Philip J Lupo, Kristin Zelle, Jaclyn Schienda, Kim E Nichols, Douglas R Stewart, David Malkin, Garrett M Brodeur, Kara Maxwell, Sharon E Plon, Michael F Walsh

1. MAI 2024 (MYPRED)

Assessment of a novel NRAS in-frame tandem duplication causing a myelodysplastic/myeloproliferative neoplasm

Exp Hematol. 2024 May. >>[Pubmed-Link](#)<<

Cora C A Beckmann, Senthilkumar Ramamoorthy, Eirini Trompouki, Wolfgang Driever, Stephan Schwarz-Furlan, Brigitte Strahm, Ayami Yoshimi, Charlotte M Niemeyer, Miriam Erlacher, Friedrich G Kapp

1. MAI 2024 (NEOCYST)

Sex, Genotype, and Liver Volume Progression as Risk of Hospitalization Determinants in Autosomal Dominant Polycystic Liver Disease

Gastroenterology. 2024 May. >>[PubMed-Link](#)<<

Ria Schönauer, Dana Sierks, Melissa Boerrigter, Tabinda Jawaid, Lea Caroff, Marie-Pierre Audrezet, Anja Friedrich, Melissa Shaw, Jan Degenhardt, Mirjam Forberger, Jonathan de Fallois, Hendrik Bläker, Carsten Bergmann, Juliana Gödiker, Philipp Schindler, Bernhard Schlevogt, Roman-U Müller, Thomas Berg, Ilse Patterson, William J Griffiths, John A Sayer; Genomics England Research Consortium; Bernt Popp, Vicente E Torres, Marie C Hogan, Stefan Somlo, Terry J Watnick, Frederik Nevens, Whitney Besse, Emilie Cornec-Le Gall, Peter C Harris, Joost P H Drenth, Jan Halbritter

1. MAI 2024 (STOP-FSGS)

The 14th International Podocyte Conference 2023: from podocyte biology to glomerular medicine

Kidney Int. 2024 May. >>[Pubmed-Link](#)<<

Sybille Koehler, Felicitas E Hengel, Bernhard Dumoulin, Laurel Damashek, Lawrence B Holzman, Katalin Susztak, Tobias B Huber; International Podocyte Conference Organizing Committee

29. APRIL 2024 (ADDRESS)

Germline biallelic BRCA2 pathogenic variants and medulloblastoma: an international cohort study

J Hematol Oncol. 2024 Apr 29. >>[Pubmed-Link](#)<<

Svenja Kastellan, Reinhard Kalb, Bia Sajjad, Lisa J McReynolds, Neelam Giri, David Samuel, Till Milde, Miriam Elbracht, Susanne Holzhauer, Marena R Niewisch, Christian P Kratz

26. APRIL 2024 (NEOCYST)

Ciliopathy patient variants reveal organelle-specific functions for TUBB4B in axonemal microtubules

Science. 2024 Apr 26. >>[Pubmed-Link](#)<<

Daniel O Dodd, et.al.

26. APRIL 2024 (TREATHSP.NET)

Activation and Purification of β -Glucocerebrosidase by Exploiting its Transporter LIMP-2 – Implications for Novel Treatment Strategies in Gaucher's and Parkinson's Disease

Adv Sci (Weinh). 2024 Apr 26. >>[PubMed-Link](#)<<

Jan Philipp Dobert, Simon Bub, Rebecca Mächtel, Dovile Janulienė, Lisa Steger, Martin Regensburger, Sibylle Wilfling, Jia-Xuan Chen, Mario Dejung, Sonja Plötz, Ute Hehr, Arne Moeller, Philipp Arnold, Friederike Zünke

24. APRIL 2024 (STOP-FSGS)

Grainyhead-like 2 Deficiency and Kidney Cyst Growth in a Mouse Model

J Am Soc Nephrol. 2024 Apr 24. >>[Pubmed-Link](#)<<

Zeliha Yesim Yurtdas, Ergin Kilic, Peter Boor, Emanuel Wyler, Markus Landthaler, Klaus Jung, Kai M Schmidt-Ott

16. APRIL 2024 (GAIN)

Truncating NFKB1 variants cause combined NLRP3 inflammasome activation and type I interferon signaling and predispose to necrotizing fasciitis

Cell Rep Med. 2024 Apr 16. >>[PubMed-Link](#)<<

Katariina Nurmi, Kristiina Silventoinen, Salla Keskitalo, Kristiina Rajamäki, Vesa-Petteri Kouri, Matias Kinnunen, Sami Jalil, Rocio Maldonado, Kirmo Wartiovaara, Elma Inés Nievas, Silvina Paola Denita-Juárez, Christopher J A Duncan, Outi Kuusmin, Janna Saarela, Inka Romo, Timi Martelius, Jukka Parantainen, Arzu Beklen, Marcelina Bilicka, Sampsa Matikainen, Dan C Nordström, Meri Kaustio, Ulla Wartiovaara-Kautto, Outi Kilpivaara, Christoph Klein, Fabian Hauck, Tiina Jahkola, Timo Hautala, Markku Varjosalo, Goncalo Barreto, Mikko R J Seppänen, Kari K Eklund

11. APRIL 2024 (STOP-FSGS)

Zyxin is important for the stability and function of podocytes, especially during mechanical stretch

Commun Biol. 2024 Apr 11. >>[PubMed-Link](#)<<

Felix Kliewe, Florian Siegerist, Elke Hammer, Jaafar Al-Hasani, Theodor Rolf Jakob Amling, Jonas Zeno Eddy Hollemann, Maximilian Schindler, Vedran Drenic, Stefan Simm, Kerstin Amann, Christoph Daniel, Maja Lindenmeyer, Markus Hecker, Uwe Völker, Nicole Endlich

9. APRIL 2024 (CONNECT-GENERATE)

Development of a Composite Score for the Clinical Assessment of Anti-IgLON5 Disease

Neurology. 2024 Apr 9. >>[PubMed-Link](#)<<

Carles Gaig, Thomas Grüter, Anna Heidbreder, Lidia Sabater, Alex Iranzo, Joan Santamaria, Frank Leypoldt, Josep O Dalmau, Ilya Ayzenberg, Francesc Graus

8. APRIL 2024 (STOP-FSGS)

Histopathological biomarkers for predicting the tumour accumulation of nanomedicines

Nat Biomed Eng. 2024 Apr 8. >>[PubMed-Link](#)<<

Jan-Niklas May, Jennifer I Moss, Florian Mueller, Susanne K Golombek, Ilaria Biancacci, Larissa Rizzo, Asmaa Said Elshafei, Felix Gremse, Robert Pola, Michal Pechar, Tomáš Etrych, Svea Becker, Christian Trautwein, Roman D Bülow, Peter Boor, Ruth Knuechel, Saskia von Stillfried, Gert Storm, Sanyogitta Puri, Simon T Barry, Volkmar Schulz, Fabian Kiessling, Marianne B Ashford, Twan Lammers

5. APRIL 2024 (STOP-FSGS)

Apoptotic cell identity induces distinct functional responses to IL-4 in efferocytic macrophages

Science. 2024 Apr 5. >>[PubMed-Link](#)<<

Imke Liebold, Amirah Al Jawazneh, Christian Casar, Clarissa Lanzloth, Stephanie Leyk, Madeleine Hamley, Milagros N Wong, Dominik Kylies, Stefanie K Gräfe, Ilka Edenhofer, Irene Aranda-Pardos, Marie Kriwet, Helmuth Haas, Jenny Krause, Alexandros Hadjilaou, Andra B Schromm, Ulricke Richardt, Petra Eggert, Dennis Tappe, Sören A Weidemann, Sourav Ghosh, Christian F Krebs, Noelia A-Gonzalez, Anna Worthmann, Ansgar W Lohse, Samuel Huber, Carla V Rothlin, Victor G Puelles, Thomas Jacobs, Nicola Gagliani, Lidia Bosurgi

1. APRIL 2024 (TREATHSP.NET)

An iPSC model for POLR3A-associated spastic ataxia: Generation of three unrelated patient cell lines

Stem Cell Res. 2024 Apr. >>[PubMed-Link](#)<<

Kalaivani Manibarathi, Tam Pham, Holger Hengel, Matthias Synofzik, Maike Nagel, Rebecca Schüle

1. APRIL 2024 (NEOCYST)

Ibrutinib-associated focal segmental glomerulosclerosis and the impact of podocin mutations in chronic lymphocytic leukemia

Kidney Int. 2024 Apr. >>[PubMed-Link](#)<<

Jan Czogalla, Simon Schliffke, Shun Lu, Maria Schwerk, Helena Petereit, Tianran Zhang, Shuya Liu, Bernhard Dumoulin, Sydney Gies, Guochao Wu, Sonja Hänzelmann, Marlies Bode, Florian Grahammer, Markus Gödel, Minna Voigtländer, Linus Butt, Carsten Bokemeyer, Carsten Bergmann, Thomas Benzing, Thorsten Wiech, Victor G Puelles, Tobias B Huber

1. APRIL 2024 (STOP-FSGS)

Ibrutinib-associated focal segmental glomerulosclerosis and the impact of podocin mutations in chronic lymphocytic leukemia

Kidney Int. 2024 Apr. >>[PubMed-Link](#)<<

Jan Czogalla, Simon Schliffke, Shun Lu, Maria Schwerk, Helena Petereit, Tianran Zhang, Shuya Liu, Bernhard Dumoulin, Sydney Gies, Guochao Wu, Sonja Hänzelmann, Marlies Bode, Florian Grahammer, Markus Gödel, Minna Voigtländer, Linus Butt, Carsten Bokemeyer, Carsten Bergmann, Thomas Benzing, Thorsten Wiech, Victor G Puelles, Tobias B Huber

20. MÄRZ 2024 (TREAT-ION)

Complex biophysical changes and reduced neuronal firing in an SCN8A variant associated with developmental delay and epilepsy

Biochim Biophys Acta Mol Basis Dis. 2024 Mar 20. >>[PubMed-Link](#)<<

Shir Quinn, Nan Zhang, Timothy A Fenton, Marina Brusel, Preethi Muruganandam, Yoav Peleg, Moshe Giladi, Yoni Haitin, Holger Lerche, Haim Bassan, Yuanyuan Liu, Roy Ben-Shalom, Moran Rubinstein

20. MÄRZ 2024 (TREATHSP.NET)

FARS-ADL across Ataxias: Construct Validity, Sensitivity to Change, and Minimal Important Change

Mov Disord. 2024 Mar 20. >>[PubMed-Link](#)<<

Andreas Traschütz, Zofia Fleszar, Holger Hengel, Thomas Klockgether, Friedrich Erdlenbruch, Björn H Falkenburger, Thomas Klopstock, Özgür Öztop-Çakmak, José Luiz Pedroso, Filippo M Santorelli, Ludger Schöls; RFC1 Study Group, PREPARE Consortium; Matthis Synofzik

15. MÄRZ 2024 (STOP-FSGS)

Lineage tracing reveals transient phenotypic adaptation of tubular cells during acute kidney injury

iScience. eCollection 2024 Mar 15. >>[PubMed-Link](#)<<

Marc Buse, Mingbo Cheng, Vera Jankowski, Michaela Lellig, Viktor Sterzer, Thiago Strieder, Katja Leuchtle, Ina V Martin, Claudia Seikrit, Paul Brinkkoettter, Giuliano Crispatsu, Jürgen Floege, Peter Boor, Timotheus Speer, Rafael Kramann, Tammo Ostendorf, Marcus J Moeller, Ivan G Costa, Eleni Stamellou

13. MÄRZ 2024 (MITONET)

Digenic Leigh syndrome on the background of the m.11778G>A Leber hereditary optic neuropathy variant

Brain. 2024 Mar 13. >>[PubMed-Link](#)<<

Beryll Blickhäuser, Sarah L Stenton, Christiane M Neuhofer, Elisa Floride, Victoria Nesbitt, Carl Fratter, Johannes Koch, Birgit Kauffmann, Claudia Catarino, Lea Dewi Schlieben, Robert Kopajtich, Valerio Carelli, Alfredo A Sadun, Robert McFarland, Fang Fang, Chiara La Morgia, Stéphanie Paquay, Marie Cécile Nassogne, Daniele Ghezzi, Costanza Lamperti, Saskia Wortmann, Jo Poulton, Thomas Klopstock, Holger Prokisch

12. MÄRZ 2024 (CONNECT-GENERATE)

[18F]DPA-714-PET-MRI reveals pronounced innate immunity in human anti-LGI1 and anti-CASPR2 limbic encephalitis

J Neurol. 2024 Mar 12. >>[PubMed-Link](#)<<

Wolfgang Roll, Jan Bauer, Andre Dik, Christoph Mueller, Philipp Backhaus, Saskia Räuber, Bastian Zinnhardt, Marco Gallus, Catriona Wimberley, Peter Körtvelyessy, Philipp Schindler, Werner Stenzel, Christian E Elger, Albert Becker, Jan Lewerenz, Heinz Wiendl, Sven G Meuth, Michael Schäfers, Nico Melzer

11. MÄRZ 2024 (NEOCYST)

Defects of renal tubular homeostasis and cystogenesis in the Pkhd1 knockout

iScience. 2024 Mar 11. >>[PubMed-Link](#)<<

Julia C Fox, Susanne T Hahnenstein, Fatima Hassan, Andrea Grund, Dieter Haffner, Wolfgang H Ziegler

8. MÄRZ 2024 (NEOCYST)

Verbesserte Versorgungs-und Behandlungsoptionen für Patienten mit Hyperphagie-assoziiierter Adipositas bei Bardet-Biedl-Syndrom [Improved Care and Treatment Options for Patients with Hyperphagia-Associated Obesity in Bardet-Biedl Syndrome]

Klin Padiatr. 2024 Mar 8. >>[PubMed-Link](#)<<

Metin Cetiner, Carsten Bergmann, Markus Bettendorf, Johanna Faust, Anja Gäckler, Bernarda Gillissen, Matthias Hansen, Maximilian Kerber, Günter Klaus, Jens König, Laura Kühlewein, Jun Oh, Annette Richter-Unruh, Julia von Schnurbein, Martin Wabitsch, Susann Weihrauch-Blüher, Lars Pape

5. MÄRZ 2024 (TREATHSP.NET)

Generation of homozygous and heterozygous REEP1 knockout induced pluripotent stem cell lines by CRISPR/Cas9 gene editing

Stem Cell Res. 2024 Mar 5. >>[PubMed-Link](#)<<

M Korneck, A Leonhardt, L Schöls, S Hauser

1. MÄRZ 2024 (HICHOL)

Odevixibat treatment in an adult patient with advanced icteric progressive cholestatic liver disease

JHEP Rep. 2023 Dec 12. eCollection 2024 Mar. >>[PubMed-Link](#)<<

Toni Herta, Carola Dröge, Adam Herber, Verena Keitel, Thomas Berg

1. MÄRZ 2024 (GAIN)

Multilayer concept of autoimmune mechanisms and manifestations in inborn errors of immunity: Relevance for precision therapy

J Allergy Clin Immunol. 2024 Mar. >>[PubMed-Link](#)<<

Markus G Seidel, Fabian Hauck

1. MÄRZ 2024 (CONNECT-GENERATE)

KCNA2 IgG autoimmunity in neuropsychiatric diseases

Brain Behav Immun. 2024 Mar. >>[PubMed-Link](#)<<

Friederike A Arlt, Ramona Miske, Marie-Luise Machule et al.

1. MÄRZ 2024 (STOP-FSGS)

Zebrafish as a model for podocyte research

Am J Physiol Renal Physiol. 2024 Mar 1. >>[PubMed-Link](#)<<

Maximilian Schindler, Nicole Endlich

1. MÄRZ 2024 (MYPRED)

Somatic variant profiling in chronic phase pediatric chronic myeloid leukemia

Haematologica. 2024 Mar 1. >>[PubMed-Link](#)<<

Yvonne Lisa Behrens, Laura Gaschler, Ronny Nienhold, Thea Reinkens, Elke Schirmer, Sabine Knöß, Renate Strasser, Stephanie Sembill, Zofia Wotschofsky, Meinolf Suttorp, Manuela Krumbholz, Brigitte Schlegelberger, Markus Metzler, Gudrun Göhring, Axel Karow

1. MÄRZ 2024 (GAIN)

Aberrant RNA sensing in regulatory T cells causes systemic autoimmunity

Sci Adv. 2024 Mar. >>[Pubmed-Link](#)<<

Domnica Luca, Sumin Lee, Keiji Hirota, Yasutaka Okabe, Junji Uehori, Kazushi Izawa, Anna-Lisa Lanz, Verena Schütte, Burcu Sivri, Yuta Tsukamoto, Fabian Hauck, Rayk Behrendt, Axel Roers, Takashi Fujita, Ryuta Nishikomori, Min Ae Lee-Kirsch, Hiroki Kato

1. MÄRZ 2024 (CONNECT-GENERATE)

Converging synaptic and network dysfunctions in distinct autoimmune encephalitis

EMBO Rep. 2024 Mar. >>[PubMed-Link](#)<<

Daniel Hunter, Mar Petit-Pedrol, Dominique Fernandes, Nathan Bénac, Catarina Rodrigues, Jakob Kreye, Mihai Ceanga, Harald Prüss, Christian Geis, Laurent Groc

28. FEBRUAR 2024 (STOP-FSGS)

Increased blood pressure after nonsevere COVID-19

J Hypertens. 2023 Nov 1. >>[PubMed-Link](#)<<

Christian Schmidt-Lauber, Elisa Alba Schmidt, Sonja Hänzelmann, Elina L Petersen, Christian-Alexander Behrendt, Raphael Twerenbold, Stefan Blankenberg, Tobias B Huber, Ulrich O Wenzel

28. FEBRUAR 2024 (MITONET)

Therapeutic benefit of idebenone in patients with Leber hereditary optic neuropathy: The LEROS nonrandomized controlled trial

Cell Rep Med. 2024 Feb 28. >>[PubMed-Link](#)<<

Patrick Yu-Wai-Man, Valerio Carelli, Nancy J Newman, Magda Joana Silva, Aki Linden, Gregory Van Stavern, Jacek P Szaflik, Rudrani Banik, Wojciech Lubiński, Berthold Pemp, Yaping Joyce Liao, Prem S Subramanian, Marta Misiuk-Hojło, Steven Newman, Lorena Castillo, Jarosław Kocięcki, Marc H Levin, Francisco Jose Muñoz-Negrete, Ali Yagan, Sylvia Cherninkova, David Katz, Audrey Meunier, Marcela Votruba, Magdalena Korwin, Jacek Dziedziak, Neringa Jurkutė, Joshua P Harvey, Chiara La Morgia, Claudia Priglinger, Xavier Llòria, Livia Tomasso, Thomas Klopstock; LEROS Study Group

21. FEBRUAR 2024 (TREATHSP.NET)

Plasma Neurofilaments: Potential Biomarkers of SPG11-Related Hereditary Spastic Paraplegia

Mov Disord. 2024 Feb 21. >>[PubMed-Link](#)<<

Laura Krumm, Jürgen Winkler, Beate Winner, Martin Regensburger

21. FEBRUAR 2024 (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

20. FEBRUAR 2024 (GENERARE)

RAS-MAPK Pathway Mutations in Congenital Pulmonary Airway Malformations

Am J Respir Crit Care Med. 2024 Feb 20. >>[PubMed-Link](#)<<

Jonas Windrich, Peter Braubach, Florian Länger, Jens Dingemann, Nicolaus Schwerk, Martin Wetzke, Diane M Renz, Martin Zenker, Denny Schanze, Christian P Kratz

20. FEBRUAR 2024 (GAIN)

Activation of gp130 signaling in T cells drives TH17-mediated multi-organ autoimmunity

Sci Signal. 2024 Feb 20. >>[PubMed-Link](#)<<

Francis Baumgartner, Stefanos A Bamopoulos, Laura Faletti, Hsiang-Jung Hsiao, Maximilian Holz, Irene Gonzalez-Menendez, Llorenç Solé-Boldo, Arik Horne, Sanket Gosavi, Ceren Özerdem, Nikita Singh, Sven Liebig, Senthilkumar Ramamoorthy, Malte Lehmann, Uta Demel, Anja A Kühl, Tim Wartewig, Jürgen Ruland, Frank T Wunderlich, Markus Schick, Wolfgang Walther, Stefan Rose-John, Simon Haas, Leticia Quintanilla-Martinez, Stefan Feske, Stephan Ehl, Rainer Glauben, Ulrich Keller

16. FEBRUAR 2024 (TREATHSP.NET)

Clinically approved immunomodulators ameliorate behavioral changes in a mouse model of hereditary spastic paraplegia type 11

Front Neurosci. 2024 Feb 16. >>[PubMed-Link](#)<<

Michaela Hörner, Sandy Popp, Julien Branchu, Giovanni Stevanin, Frédéric Darios, Stephan Klebe, Janos Groh, Rudolf Martini

13. FEBRUAR 2024 (TREATHSP.NET)

Recommendations for optimal interdisciplinary management and healthcare settings for patients with rare neurological diseases

Orphanet J Rare Dis. 2024 Feb 13. >>[PubMed-Link](#)<<

Holm Graessner, Carola Reinhard, Tobias Bäumer, Annette Baumgärtner, Knut Brockmann, Norbert Brüggemann, Eva Bültmann, Jeanette Erdmann, Kirstin Heise, Günter Höglinger, Irina Hüning, Frank J Kaiser, Christine Klein, Thomas Klopstock, Ingeborg Krägeloh-Mann, Markus Kraemer, Kerstin Luedtke, Martin Mücke, Thomas Musacchio, Andreas Nadke, Alma Osmanovic, Gabriele Ritter, Katharina Röse, Christopher Schippers, Ludger Schöls, Rebecca Schüle, Jörg B Schulz, Joachim Sproß, Eveline Stasch, Gilbert Wunderlich, Alexander Münchau

2. FEBRUAR 2024 (TREATHSP.NET)

Neuroinflammatory disease signatures in SPG11-related hereditary spastic paraplegia patients

Acta Neuropathol. 2024 Feb 2. >>[PubMed-Link](#)<<

Laura Krumm, Tatyana Pozner, Naime Zagha, Roland Coras, Philipp Arnold, Thanos Tsaktanis, Kathryn Scherpelz, Marie Y Davis, Johanna Kaindl, Iris Stolzer, Patrick Süß, Mukhran Khundadze, Christian A Hübner, Markus J Riemenschneider, Jonathan Baets, Claudia Günther, Suman Jayadev, Veit Rothhammer, Florian Krach, Jürgen Winkler, Beate Winner, Martin Regensburger

1. FEBRUAR 2024 (STOP-FSGS)

Inhibition of the renal apical sodium dependent bile acid transporter prevents cholemic nephropathy in mice with obstructive cholestasis

J Hepatol. 2024 Feb. >>[PubMed-Link](#)<<

Ahmed Ghallab, et.al.

1. FEBRUAR 2024 (MYPRED)

Clonal hematopoiesis in children with predisposing conditions

Semin Hematol. 2024 Feb. >>[PubMed-Link](#)<<

Enrico Attardi, Seth J Corey, Marcin W Wlodarski

1. FEBRUAR 2024 (MYPRED)

Spontaneous remission and loss of monosomy 7: a window of opportunity for young children with SAMD9L syndrome

Haematologica. 2024 Feb 1. >>[PubMed-Link](#)<<

Miriam Erlacher, Felicia Andresen, Martina Sukova, Jan Sary, Barbara De Moerloose, Jutte van der Werff Ten Bosch, Michael Dworzak, Markus G Seidel, Sophia Polychronopoulou, Rita Beier, Christian P Kratz, Michaela Nathrath, Michael C Frühwald, Gudrun Göhring, Anke K Bergmann, Christina Mayerhofer, Dirk Lebrecht, Senthilkumar Ramamoorthy, Ayami Yoshimi, Brigitte Strahm, Marcin W Wlodarski, Charlotte M Niemeyer

1. FEBRUAR 2024 (MYPRED)

Biallelic inactivation of the NF1 tumour suppressor gene in juvenile myelomonocytic leukaemia: Genetic evidence of driver function and implications for diagnostic workup

Br J Haematol. 2024 Feb. >>[PubMed-Link](#)<<

Senthilkumar Ramamoorthy, Dirk Lebrecht, Denny Schanze, Ina Schanze, Ilse Wieland, Geoffroy Andrieux, Patrick Metzger, Maria Hess, Michael H Albert, Arndt Borkhardt, Dorine Bresters, Jochen Buechner, Albert Catala, Valerie De Haas, Michael Dworzak, Miriam Erlacher, Henrik Hasle, Kirsi Jahnukainen, Franco Locatelli, Riccardo Masetti, Jan Sary, Dominik Turkiewicz, Luca Vinci, Marcin W Wlodarski, Ayami Yoshimi, Melanie Boerries, Charlotte M Niemeyer, Martin Zenker, Christian Flothozik

27. JANUAR 2024 (STOP-FSGS)

The podocytes' inflammatory responses in experimental GN are independent of canonical MYD88-dependent toll-like receptor signaling

Sci Rep. 2024 Jan 27. >>[PubMed-Link](#)<<

Thomas Schömig, Paul Diefenhardt, Ingo Plagmann, Bastian Trinsch, Tim Merz, Giuliano Crispatzu, David Unnersjö-Jess, Jasper Nies, David Pütz, Claudio Sierra Gonzalez, Bernhard Schermer, Thomas Benzing, Paul Thomas Brinkkoetter, Sebastian Brähler

17. JANUAR 2024 (MITONET)

Genetic variants affecting NQO1 protein levels impact the efficacy of idebenone treatment in Leber hereditary optic neuropathy

Cell Rep Med. 2024 Jan 17. >>[PubMed-Link](#)<<

Serena Jasmine Aleo, Valentina Del Dotto, Martina Romagnoli, Claudio Fiorini, Giada Capirossi, Camille Peron, Alessandra Maresca, Leonardo Caporali, Mariantonietta Capristo, Concetta Valentina Tropeano, Claudia Zanna, Fred N Ross-Cisneros, Alfredo A Sadun, Maria Gemma Pignataro, Carla Giordano, Chiara Fasano, Andrea Cavaliere, Anna Maria Porcelli, Gaia Tioli, Francesco Musiani, Alessia Catania, Costanza Lamperti, Stefania Bianchi Marzoli, Annamaria De Negri, Maria Lucia Cascavilla, Marco Battista, Piero Barboni, Michele Carbonelli, Giulia Amore, Chiara La Morgia, Dmitrii Smirnov, Catalina Vasilescu, Aiman Farzeen, Beryll Blickhaeuser, Holger Prokisch, Claudia Priglinger, Bettina Livonius, Claudia B Catarino, Thomas Klopstock, Valeria Tiranti, Valerio Carelli, Anna Maria Ghelli

11. JANUAR 2024 (GAIN)

Disrupted degradative sorting of TLR7 is associated with human lupus

Sci Immunol. 2024 Jan 11. >>[PubMed-Link](#)<<

Harshita Mishra, Claire Schlack-Leigers, Ee Lyn Lim, Oliver Thieck, Thomas Magg, Johannes Raedler, Christine Wolf, Christoph Klein, Helge Ewers, Min Ae Lee-Kirsch, David Meierhofer, Fabian Hauck, Olivia Majer

11. JANUAR 2024 (GAIN)

UNC93B1 variants underlie TLR7-dependent autoimmunity

Sci Immunol. 2024 Jan 11. >>[PubMed-Link](#)<<

Christine Wolf, Ee Lyn Lim, Mohammad Mokhtari, Barbara Kind, Alexandru Odainic, Eusebia Lara-Villacanas, Sarah Koss, Simon Mages, Katharina Menzel, Kerstin Engel, Gregor Dückers, Benedikt Bernbeck, Dominik T Schneider, Kathrin Siepermann, Tim Niehues, Carl Christoph Goetzke, Pawel Durek, Kirsten Minden, Thomas Dörner, Anna Stittrich, Franziska Szelinski, Gabriela Maria Guerra, Mona Massoud, Markus Bieringer, Carina C de Oliveira Mann, Eduardo Beltrán, Tilmann Kallinich, Mir-Farzin Mashreghi, Susanne V Schmidt, Eicke Latz, Johanna Klughammer, Olivia Majer, Min Ae Lee-Kirsch

1. JANUAR 2024 (GAIN)

Autoimmune lymphoproliferative immunodeficiencies (ALPIDs): A proposed approach to redefining ALPS and other lymphoproliferative immune disorders

J Allergy Clin Immunol. 2024 Jan. >>[PubMed-Link](#)<<

Aude Magerus, Anne Rensing-Ehl, V Koneti Rao, David T Teachey, Frederic Rieux-Laucat, Stephan Ehl

1. JANUAR 2024 (GAIN)

PLCG2-associated immune dysregulation (PLAID) comprises broad and distinct clinical presentations related to functional classes of genetic variants

J Allergy Clin Immunol. 2024 Jan. >>[PubMed-Link](#)<<

Kathleen Baysac, Guangping Sun, Hiroto Nakano, Elizabeth G Schmitz, Anthony C Cruz, Charles Fisher, Alexis C Bailey; PLCG2-Immune Dysregulation Working Group; Emily Mace, Joshua D Milner, Michael J Ombrello

1. JANUAR 2024 (TREATHSP.NET)

Clinical and genetic keys to cerebellar ataxia due to FGF14 GAA expansions

EBioMedicine. 2024 Jan. >>[PubMed-Link](#)<<

Jean-Loup Méreaux, Claire-Sophie Davoine, David Pellerin, Giulia Coarelli, Marie Coutelier, Claire Ewencyk, Marie-Lorraine Monin, Mathieu Anheim, Isabelle Le Ber, Stéphane Thobois, Florent Gobert, Léna Guillot-Noël, Sylvie Forlani, Ludmila Jornea, Anna Heinzmann, Aude Sangare, Bertrand Gaymard, Lucie Guyant-Maréchal, Perrine Charles, Cecilia Marelli, Jérôme Honnorat, Bertrand Degos, François Tison, Sophie Sangla, Marion Simonetta-Moreau, François Salachas, Maya Tchikviladze, Giovanni Castelnovo, Fanny Mochel, Stephan Klebe, Anna Castrioto, Silvia Fenu, Aurélie Méneret, Frédéric Bourdain, Marion Wandzel, Virginie Roth, Céline Bonnet, Florence Riant, Giovanni Stevanin, Sandrine Noël, Anne-Laure Fauret-Amsellem, Melanie Bahlo, Paul J Lockhart, Bernard Brais, Mathilde Renaud, Alexis Brice, Alexandra Durr

1. JANUAR 2024 (GAIN)

Hypogammaglobulinemia and immune dysregulation-not just 2 sides of a coin

J Allergy Clin Immunol. 2024 Jan. >>[PubMed-Link](#)<<

Gulbu Uzel, Baerbel Keller, Klaus Warnatz

1. JANUAR 2024 (CONNECT-GENERATE)

Early Neuroaxonal Damage in Neurologic Disorders Associated With GAD65 Antibodies

Neurol Neuroimmunol Neuroinflamm. 2023 Nov 1. Print 2024 Jan. >>*Pubmed-Link*<<

Katharina Eisenhut, Jennifer Faber, Daniel Engels, Ramona Gerhards, Jan Lewerenz, Kathrin Doppler, Claudia Sommer, Robert Markewitz, Kim K Falk, Rosa Rössling, Harald Pruess, Carsten Finke, Jonathan Wickel, Christian Geis, Dominica Ratuszny, Lena K Pfeffer, Stefan Bittner, Johannes Piepgras, Andrea Kraft, Jaqueline Klausewitz, Brigitte Nuscher, Tania Kümpfel, Franziska S Thaler; German Network for Research on Autoimmune Encephalitis (GENERATE)