

Veröffentlichungen der BMFTR-geförderten Forschungsverbünde für Seltene Erkrankungen, 2025

31. DEZEMBER 2025 (STOP-FSGS)

Disruption of Mitochondrial Dynamics and Integrity Drives Divergent Metabolic Flexibility and Resilience in Podocytes

FASEB J. 2025 Dec 31. >>[Pubmed-Link](#)<<

Cem Özel, Katrin M Reitmeier, Emilia Kieckhöfer, Khawla Abualia, Duc Nguyen-Minh, Mahsa Matin, Henning Hagmann, Richard J M Coward, Sebastian Brähler, Philipp Antczak, Bernhard Schermer, Thomas Benzing, Patrick Gialvalisco, Paul T Brinkkoetter

23. DEZEMBER 2025 (CONNECT-GENERATE)

Secondary obsessive-compulsive syndromes: a systematic literature review resulting in 228 suspected cases

Mol Psychiatry. 2025 Dec 23. >>[Pubmed-Link](#)<<

Kim Runge, Bernd Feige, Miriam A Schiel, Katharina von Zedtwitz, Alexander Maier, Nils Stöcker, Raphael J Dressle, Juan C Baldermann, Simon J Maier, Kathrin Nickel, Harald Prüss, Volker A Coenen, Ulrich Voderholzer, Katharina Domschke, Ludger Tebartz van Elst, Dominique Endres

14. DEZEMBER 2025 (MYPRED)

Concurrent Juvenile Myelomonocytic Leukemia and Gliomas in Patients With Neurofibromatosis Type 1

Pediatr Blood Cancer. 2025 Dec 27. >>[Pubmed-Link](#)<<

Henry Hongo, Sarah K Tasian, Alix E Seif, Jenna Rossoff, Alicia Lenzen, Barbara Spitzer, Michael Dworzak, Claas Röhl, Roland Meisel, Marianne Hoffmann, Elliot Stieglitz, Christian Flotho

12. DEZEMBER 2025 (STOP-FSGS)

Tumor-stromal crosstalk and macrophage enrichment are associated with chemotherapy response in bladder cancer

FEBS Open Bio. 2025 Dec 12. >>[Pubmed-Link](#)<<

Sophie Leybold, Janik Riese, Lancelot Seillier, Mark Kühnel, Julia Pannhausen, Charlotte O J Fröhlich, Christian Martin, Peter Boor, Matthias Saar, Danny D Jonigk, Nadine T Gaisa, Michael Rose

11. DEZEMBER 2025 (MYPRED)

RUNX1-FPDMM in families with mild thrombocytopenia and platelet function anomalies: a case series

Front Med (Lausanne). 2025 Dec 11. >>[Pubmed-Link](#)<<

Hannah Glonnegger, Doris Boeckelmann, Rebekka Wiedenhöfer, Wolf-Achim Hassenpflug, Tim Ripperger, Dirk Lebrecht, Ralf Knöfler, Oliver Tiebel, Udo Koehler, Claudia Wehr, Harry Sirb, Monika Sparber-Sauer, Katrin Reinsberger, Ayami Yoshimi, Brigitte Strahm, Barbara Zieger

9. DEZEMBER 2025 (CONNECT-GENERATE)

Anti-NMDA-Receptor GluN1 Antibody Serostatus Is Robust in Acute Severe Stroke

Diagnostics (Basel). 2025 Dec 9. >>[Pubmed-Link](#)<<

Pia Sophie Sperber, Benjamin Hotter, Matthias Endres, Harald Prüss, Andreas Meisel

9. DEZEMBER 2025 (ADDRESS)

Neoantigenic properties of TP53 variants influence cancer risk in individuals with Li-Fraumeni syndrome

EBioMedicine. 2025 Dec 9. >>[Pubmed-Link](#)<<

Emilie Montellier, Olivier Manches, Jonathan Gaucher, Claire Freycon, David Hoyos, Sandrine Blanchet, Murielle Verboom, Christina M Dutzmann, Sophie Coutant, Jacqueline Bou, Bertrand Fin, Robert Olaso, Jean-François Deleuze, Thierry Frébourg, Benjamin D Greenbaum, Arnold J Levine, Christian P Kratz, Gaëlle Bougeard, Pierre Hainaut

9. DEZEMBER 2025 (HICHOL)

Uncovering differences in rye and wheat degradation by human gut microbiota applying a quantitative multi-metaOmics in vitro approach

Curr Res Microb Sci. 2025 Dec 9. eCollection 2026. >>[Pubmed-Link](#)<<

Clara Berenike Arnold, Alexandra Kelder, Sabrina Woltemate, Robert Geffers, Andreas von Felde, Knud Erik Bach Knudsen, Christian Visscher, Marius Vital

4. DEZEMBER 2025 (TREATHSP.NET)

Feasibility of a smartphone application for remote use in spastic ataxias: an 8-week long-term PROSPAX study

J Neurol. 2025 Dec 4. >>[Pubmed-Link](#)<<

Ilse H J Willemse, Sabato Mellone, Carlo Tacconi, Filippo M Santorelli, Ivana Ricca, Sara Satolli, Stephan Klebe, Nicole Jeschonneck, Winfried Ilg, Rebecca Schüle, Matthis Synofzik, Jorik Nonnekes, Bart P C van de Warrenburg

1. DEZEMBER 2025 (STOP-FSGS)

Patient anti-PLA2R1 autoantibodies cause membranous nephropathy in human PLA2R1 transgenic mice

Kidney Int. 2025 Dec. >>[Pubmed-Link](#)<<

Gunther Zahner, Silke Dehde, Larissa Seifert, Ming Huang, Oliver Kretz, Tobias B Huber, Nicola M Tomas

1. DEZEMBER 2025 (ADDRESS)

Psychosocial burdens and unmet supportive care needs of partners and relatives of individuals with Li-Fraumeni syndrome: A mixed-methods study

J Genet Couns. 2025 Dec. >>[Pubmed-Link](#)<<

Senta Kiermeier, Sarah Schott, Juliane Nees, Christina M Dutzmann, Farina Silchmüller, Christian P Kratz, Myriam Keymling, Imad Maatouk

1. DEZEMBER 2025 (STOP-FSGS)

Advances in computational nephropathology

Kidney Int. 2025 Dec. >>[Pubmed-Link](#)<<

David L Hölscher, Roman D Bülow, Martin Strauch, Peter Boor

30. NOVEMBER 2025 (STOP-FSGS)

Kynurenine Pathway Dysregulation Impairs Podocyte Morphology and Bioenergetics In Vitro and Leads to Glomerular Dysfunction

FASEB J. 2025 Nov 30. >>[Pubmed-Link](#)<<

Patricia Bolanos-Palmieri, Heiko Schenk, Heike Bähre, Patricia Schroder, Lynne Staggs, Hermann Haller, Mario Schiffer

28. NOVEMBER 2025 (STOP-FSGS)

Arp2/3-dependent regulation of ciliogenesis governs adaptive distal tubular epithelial cell states in kidney disease

Sci Adv. 2025 Nov 28. >>[Pubmed-Link](#)<<

Manuel Rogg, Lisa Weißer, Jasmin I Maier, August Sigle, Martin Helmstädter, Marlene Stigler, Alena Sammarco, Katja Gräwe, Grigor Andreev, Charlotte Kark, Suresh K Ramakrishnan, Cem Özel, Linus Butt, Frederic Arnold, Wibke Bechtel-Walz, Oliver Schilling, Yakup Tanriver, Paul Brinkkötter, Markus Grabbert, Matias Simons, Martin Werner, Oliver Kretz, Thomas Benzing, Tobias B Huber, Christoph Schell

27. NOVEMBER 2025 (GAIN)

Do CVID patients on SCIG have more autoimmune (thrombo)cytopenic events than CVID patients on IVIG?

Front Immunol. 2025 Nov 27. >>[Pubmed-Link](#)<<

Nadezhda Camacho-Ordonez, Aleksandra Hirsch, Luiza Campos, Sigune Goldacker, Siobhan O Burns, Fernando Moreira, Klaus Warnatz, Bodo Grimbacher

20. NOVEMBER 2025 (STOP-FSGS)

A systematic analysis of the impact of data variation on AI-based histopathological grading of prostate cancer

Med Image Anal. 2026 Feb. Epub 2025 Nov 20 >>[Pubmed-Link](#)<<

Patrick Fuhlert, Fabian Westhaeusser, Esther Dietrich, Maximilian Lennartz, Robin Khatri, Nico Kaiser, Pontus Röbeck, Roman Bülow, Saskia Von Stillfried, Anja Witte, Sam Ladjevardi, Anders Drotte, Peter Severgårdh, Jan Baumbach, Victor G Puellas, Michael Häggman, Michael Brehler, Peter Boor, Peter Walhagen, Anca Dragomir, Christer Busch, Markus Graefen, Ewert Bengtsson, Guido Sauter, Marina Zimmermann, Stefan Bonn

15. NOVEMBER 2025 (CONNECT-GENERATE)

Heterogeneity of anti-Caspr2 antibodies: specificity and pathogenicity

Transl Psychiatry. 2025 Nov 15. >>[Pubmed-Link](#)<<

Julia Su, Rohan Gupta, Scott van Hoof, Jakob Kreye, Harald Prüß, Benjamin Spielman, Lior Brimberg, Bruce T Volpe, Patricio T Huerta, Betty Diamond

14. NOVEMBER 2025 (MITONET)

The genotypic and phenotypic landscape of PDHA1-related pyruvate dehydrogenase complex deficiency
Brain. 2025 Nov 14. >>[Pubmed-Link](#)<<

Kajus Merkevicius, (...), Thomas Klopstock (...), Martin Zenker, (...), Holger Prokisch, Johannes A Mayr, Saskia B Wortmann

5. NOVEMBER 2025 (MYPRED)

Tuning of G-CSFR signaling by de novo-designed agonists

Mol Ther. 2025 Nov 5. >>[Pubmed-Link](#)<<

Timo Ullrich, Christoph Pollmann, Malte Ritter, Jérémy Haaf, Narges Aghaallaei, Ivan Tesakov, Valeriia Hatskovska, Maya El-Riz, Kateryna Maksymenko, Sergey Kandabarau, Maksim Klimiankou, Claudia Lengerke, Karl Welte, Birte Hernandez-Alvarez, Patrick Müller, Andrei Lupas, Jacob Piehler, Julia Skokowa, Mohammad ElGamacy

5. NOVEMBER 2025 (GAIN)

Compound heterozygous NFKB1 missense variants in cis associated with immunodeficiency

Clin Immunol. 2026 Jan. Epub 2025 Nov 5 >>[Pubmed-Link](#)<<

Manfred Fliegau, Kelly Nitschke, Pavla Mrovecova, Sara Posadas-Cantera, Nadezhda Camacho-Ordóñez, Klaus Warnatz, Baerbel Keller, Bodo Grimbacher

1. NOVEMBER 2025 (TREAT-ION)

Modeling AP2M1 developmental and epileptic encephalopathy in Drosophila

Dis Model Mech. 2025 Nov 1. >>[Pubmed-Link](#)<<

Robin A Karge, Florian P Fischer, Hannah Schüth, Aileen Wechner, Sabrina Peter, Lukas A Kilo, Mato Dichter, Aaron Voigt, Gaia Tavasani, Karen M J van Loo, Henner Koch, Yvonne G Weber, Stefan Wolking

1. NOVEMBER 2025 (CONNECT-GENERATE)

The Neuropsychiatric Checklist for Autoimmune Psychosis: A Narrative Review

Biol Psychiatry. 2025 Nov 1. >>[Pubmed-Link](#)<<

Ludger Tebartz van Elst, Kimon Runge, Philipp T Meyer, Horst Urbach, Nils Venhoff, Harald Prüss

1. NOVEMBER 2025 (STOP-FSGS)

Deletion of ABIN1-LIR motifs impairs hepatic lipid homeostasis and mitophagy via AMPK-TFEB axis in mice

Am J Physiol Cell Physiol. 2025 Nov 1. >>[Pubmed-Link](#)<<

Lisa Sophie Huber, Rosetta Merline, Jinyang Zeng-Brouwers, Rajkumar Vutukuri, Nico Kraus, Cristina Ortiz, Stefan Guenther, Eva Miriam Buhl, Lisa Hahnefeld, Robert Gurke, Julia Bein, Madina Karimova, Patrick Wurzel, Peter Boor, Christoph Welsch, Peter Wild, Josef Pfeilschifter, Donat Kögel, Malgorzata Wygrecka, Jonel Trebicka, Rafal Bartoszewski, Ivan Dikic, Liliana Schaefer

1. NOVEMBER 2025 (TREATHSP.NET)

Hyperactivity of the non-canonical inflammasome in SPG11 and SPG48

EBioMedicine. 2025 Nov. >>[Pubmed-Link](#)<<

Muhammad Awais Afzal, Mohamed Ghait, Adeela Hussain, Anke Siegmund, Lorena Tuchscher, Petra Babic, Adrian T Press, Robert Hardt, Dominic Winter, Annekathrin Rödiger, Rebecca Schüle, Jens Fielitz, Michael Bauer, Christian Andreas Hübner

1. NOVEMBER 2025 (STOP-FSGS)

BioContextAI is a community hub for agentic biomedical systems

Nat Biotechnol. 2025 Nov. >>[Pubmed-Link](#)<<

Malte Kuehl, Darius P Schaub, Francesco Carli, Lukas Heumos, Malte Hellmig, Camila Fernández-Zapata, Nico Kaiser, Jonathan Schaul, Anton Kulaga, Nikolay Usanov; Scverse Community; Christian F Krebs, Ulf Panzer, Stefan Bonn, Sebastian Lobentanz, Julio Saez-Rodriguez, Victor G Puelles

1. NOVEMBER 2025 (HICHOL)

A farnesoid X receptor T296I variant disrupts ligand-induced FXR activation and thus bile acid transport in progressive familial intrahepatic cholestasis

J Biol Chem. 2025 Nov. >>[Pubmed-Link](#)<<

Annika Behrendt, Alex Bastianelli, Jan Stindt, Eva-Doreen Pfister, Malte Sgodda, Tobias Cantz, Sebastian Hook, Mohanraj Gopalswamy, Kathrin Grau, Stefanie Brands, Carola Dröge, Amelie Stalke, Michele Bonus, Sabine Franke, Ulrich Baumann, Verena Keitel, Holger Gohlke

10. OKTOBER 2025 (CONNECT-GENERATE)

Binding of established antinuclear antibodies to neurons depends on tissue fixation and underlying autoantigens

Front Immunol. 2025 Oct 10. >>[Pubmed-Link](#)<<

Lucie Y Li, Markus Höltje, Helle Foverskov Rasmussen, Lennard Halle, Marie Mayrhofer, Martin Blüthner, Harald Prüss

1. OKTOBER 2025 (HICHOL)

iPSC-based hepatic organoids reveal a heterozygous MYO5B variant as driver of intrahepatic cholestasis

Hepatol Commun. 2025 Sep 29. eCollection 2025 Oct 1. >>[Pubmed-Link](#)<<

Malte Sgodda, Evelyn Gebel, Lennart Dignas, Susanne Alfken, Reto Eggenschwiler, Amelie Stalke, Carola Dröge, Eva-Doreen Pfister, Ulrich Baumann, Tom Luedde, Irene Esposito, Verena Keitel, Tobias Cantz

1. OKTOBER 2025 (TREATHSP.NET)

Deciphering brain organoid heterogeneity by identifying key quality determinants

Commun Biol. 2025 Oct 1. >>[Pubmed-Link](#)<<

Tom Boerstler, Daniil Kachkin, Elizaveta Gerasimova, Naime Zagha, Federica Furlanetto, Negar Nayebezhade, Luke Zappia, Michelle Boisvert, Michaela Farrell, Sonja Ploetz, Iryna Prots, Martin Regensburger, Claudia Günther, Juergen Winkler, Pooja Gupta, Fabian Theis, Marisa Karow, Sven Falk, Beate Winner, Florian Krach

29. SEPTEMBER 2025 (ADDRESS)

TP53 variant clusters stratify phenotypic diversity in germline carriers and reveal an osteosarcoma-prone subgroup

Nat Commun. 2025 Sep 29. >>[Pubmed-Link](#)<<

Nicholas W Fischer, Noel Ong, Brianne Laverty, Pamela Psarianos, Camilla Giovino, Noa Alon, Emilie Montellier, Pierre Hainaut, Kara N Maxwell, Christian P Kratz, Ran Kafri, David Malkin

23. SEPTEMBER 2025 (STOP-FSGS)

Metabolite-enhanced normothermic machine perfusion improves kidney transplant viability

JCI Insight. 2025 Sep 23. >>[Pubmed-Link](#)<<

Jan Czogalla, Fabian Hausmann, Simon Lagies, Sydney E Gies, Sabrina Christiansen, Nico Kaiser, Fabian Haas, Yusuke Okabayashi, Dominik Kyles, Smilla Hofmann, Rossana Franzin, Niklas Sabra, Sarah Bouari, Yitian Fang, Gisela Ambagtsheer, Ilka Edenhofer, Silvia Chilla, Anne K Mühlig, Marina Zimmermann, Milagros N Wong, Takashi Yokoo, Oliver Kretz, Maja Lindenmeyer, Florian Grahammer, Martin J Hoogduijn, Ron de Bruin, Malte Kuehl, Sonja Hänzelmann, Bernd Kammerer, Loreto Gesualdo, Stefan Bonn, Robert C Minnee, Tobias B Huber, Victor G Puelles

17. SEPTEMBER 2025 (CONNECT-GENERATE)

MR spectroscopic imaging and its association with EEG, CSF, and psychometric/neuropsychological findings in patients with suspected autoimmune psychosis spectrum syndromes

Acta Neuropsychiatr. 2025 Sep 17. >>[Pubmed-Link](#)<<

Dominique Endres, Isabelle Matteit, Katharina von Zedtwitz, Bernd Feige, Andrea Schlump, Marco Reisert, Kathrin Nickel, Kimon Runge, Katharina Domschke, Evgeniy Perlov, Alexander Rau, Harald Prüss, Thomas Lange, Ludger Tebartz van Elst, Simon J Maier

17. SEPTEMBER 2025 (TREATHSP.NET)

Serum NfL, but not GFAP, differentiates primary lateral sclerosis from adrenomyeloneuropathy and hereditary spastic paraplegia type 4

Amyotroph Lateral Scler Frontotemporal Degener. 2025 Sep 17. >>[Pubmed-Link](#)<<

Christoph Kessler, Carlo Wilke, Holger Hengel, Tim W Rattay, Aleksandra Maleska Macieski, Jens Kuhle, Ludger Schöls, Rebecca Schüle

15. SEPTEMBER 2025 (CONNECT-GENERATE)

Risk of Epilepsy and Factors Associated With Time to Seizure Remission in Anti-LGI1 Encephalitis: Long-Term Outcome in 236 Patients

Neurol Neuroimmunol Neuroinflamm. 2025 Nov. Epub 2025 Sep 15. >>[Pubmed-Link](#)<<

Tobias Baumgartner, Moritz Freyberg, Lucia Campetella, Yvette Crijnen, Justina Dargvainiene, Charlotte Behning, Christian G Bien, Anna Rada, Harald Prüss, Rosa Rössling, Stjepana Kovac, Christine Strippel, Franziska S Thaler, Katharina Eisenhut, Jan Lewerenz, Felicitas Becker, Raphael Reinecke, Michael Peter Malter, Kurt-Wolfram Sühs, Simone C Tauber, Felix Von Podewils, Nico Melzer, Klaus-Peter Wandinger, Romina-Anna-Maria Fernandez Ceballos, Jens Kuhle, Klaus Berger, Tobias Bauer, Theodor Rüber, Attila Racz, Albert J Becker, Julika Pitsch, Gregor Kuhlenbäumer, Sergio Muñoz-Castrillo, Jerome Honnorat, Maarten J Titulaer, Frank Leypoldt, Rainer Surges; and the GENERATE study group

12. SEPTEMBER 2025 (NEOCYST)

Tulp3 deficiency results in ciliopathy phenotypes during zebrafish embryogenesis

Sci Rep. 2025 Sep 12. >>[Pubmed-Link](#)<<

Daniel Epting John Devane, Ralf Mertes, Séverine Kayser, Martin Helmstädter, Patrick Metzger, Melanie Boerries, Carsten Bergmann, Elisabeth Ott

9. SEPTEMBER 2025 (NEOCYST)

PATJ deficiency leads to cystic kidney disease and related ciliopathies

HGG Adv. 2025 Sep 9. >>[Pubmed-Link](#)<<

Daniel Epting, Daniela A Braun, Eva Decker, Elisabeth Ott, Tobias Eisenberger, Nadine Bachmann, Pavel Nedvetsky, Michael P Krahn, Friedhelm Hildebrandt, Carsten Bergmann

3. SEPTEMBER 2025 (MITONET)

Repeat-associated ataxias in a German patient cohort analysed by targeted parallel long-read sequencing

Brain. 2025 Sep 3. >>[Pubmed-Link](#)<<

Hannes Erdmann, Annalisa Schaub, Morghan C Lucas, Veronika Scholz, Anna Benet-Pages, Kerstin Becker, Christine Dineiger, Veronika Mayer, Inga van Buren, Eva Breithausen, Karl Akbari, Isabell Cordts, Mayra Sauer, Christine Schneider, Rosanna Krakowsky, Franziska Schnabel, Konstanze Dunker, Lena Fabritius, Johannes Gerb, Denis Grabova, Ken Möhwald, Marius Näher, Karoline Steinmetz, Franziska Thiessen, Alexander Jäck, Christiane Schneider-Gold, Simone Zittel, Christina Petersen, Isolde Schreyer, Larissa Mämecke, Sibylle Wilfling, Gilbert Wunderlich, David Brenner, Yorck Hellenbroich, Kirsten Muhle, Tessa Huchtemann, Inga Claus, Thomas Klopstock, Michael Strupp, Johannes Levin, Günter Höglinger, Doreen Huppert, Sandra Becker-Bense, Philipp Filippopoulos, Fabian Kilpert, Elsa Leitão, Sabine Kaya, Christel Depienne, Florian Schöberl, Teresa Neuhaus, Elke Holinski-Feder, Andreas Zwergal, Angela Abicht

1. SEPTEMBER 2025 (HICHOL)

Circulating and faecal microRNAs as non-invasive biomarkers for IBD: current evidence and next steps

Best Pract Res Clin Gastroenterol. 2025 Sep. >>[Pubmed-Link](#)<<

Vytautas Kiudelis, Juozas Kupcinskis, Alexander Link

1. SEPTEMBER 2025 (ADDRESS)

Model of care for individuals with rare cancer predisposition syndromes in Germany

The Lancet Regional Health – Europe. Sept. 2025. >>[Journal-Link](#)<<

Valentina Härter, Birte Sängers, Josephine C. Giesecke, Tanja Gerasimov, Beatrice Hoffmann, Carolin Huisinga, Anja Karow, Lucas J. Müntnich, Natalie E. Palmaers, Stefanie Paquet, Judith Penkert, Diane M. Renz, Tim Ripberger, Farina J. Silchmüller, Christina M. Dutzmann, Christian P. Kratz

1. SEPTEMBER 2025 (NEOCYST)

Polycystic Kidney Disease in Children: The Current Status and the Next Horizon

Am J Kidney Dis. 2025 Sep. >>[Pubmed-Link](#)<<

Melissa A Cadnapaphornchai, Katherine M Dell, Charlotte Gimpel, Lisa M Guay-Woodford, Ashima Gulati, Erum A Hartung, Max C Liebau, Andrew J Mallett, Matko Marlais, Djalila Mekahli, Alixandra Piccirilli, Tomas Seeman, Kristin Tindal, Paul J D Winyard

1. SEPTEMBER 2025 (MYPRED)

Model of care for individuals with rare cancer predisposition syndromes in Germany

The Lancet Regional Health – Europe. Sept. 2025. >>[Journal-Link](#)<<

Valentina Härter, Birte Sängler, Josephine C. Gieseke, Tanja Gerasimov, Beatrice Hoffmann, Carolin Huisinga, Anja Karow, Lucas J. Müntnich, Natalie E. Palmaers, Stefanie Paquet, Judith Penkert, Diane M. Renz, Tim Ripperger, Farina J. Silchmüller, Christina M. Dutzmann, Christian P. Kratz

1. SEPTEMBER 2025 (ADDRESS)

scafari: exploring scDNA-seq data

Bioinformatics. 2025 Sep 1. >>[Pubmed-Link](#)<<

Sophie-Marie Wind, Thea Reinkens, Yvonne Lisa Behrens, Sarah Sandmann

1. SEPTEMBER 2025 (MYPRED)

scafari: exploring scDNA-seq data

Bioinformatics. 2025 Sep 1. >>[Pubmed-Link](#)<<

Sophie-Marie Wind, Thea Reinkens, Yvonne Lisa Behrens, Sarah Sandmann

29. AUGUST 2025 (GAIN)

STAT3 haploinsufficiency is associated with autosomal dominant hyper-IgE syndrome

Sci Adv. 2025 Aug 29. >>[Pubmed-Link](#)<<

Virginia Andreani, Aaron James Forde, Manfred Fliegauf, Giulia Bressan, Vera Noé, Nils Ott, Shiva Saghafi, Larsen Vornholz, Sophie E Isay, Jürgen Ruland, Philipp Henneke, Bodo Grimbacher

28. AUGUST 2025 (STOP-FSGS)

Platelet-derived growth factor-C contributes to kidney inflammation in experimental hypertension with little effect on the peritubular capillary network

Exp Mol Pathol. 2025 Aug 28. >>[Pubmed-Link](#)<<

Ina V Martin, Christian Dippel, Eva M Buhl, Robert Göllinger, Katja Ermert, Jürgen Floege, Eleni Stamellou, Ute Raffetseder, Rafael Kramann, Tammo Ostendorf

26. AUGUST 2025 (MITONET)

RTN4IP1 is required for the final stages of mitochondrial complex I assembly and CoQ biosynthesis

EMBO J. 2025 Aug 26. >>[Pubmed-Link](#)<<

Monika Oláhová, Rachel M Guerra, Jack J Collier, Juliana Heidler, Kyle Thompson, Chelsea R White, Paulina Castañeda-Tamez, Alfredo Cabrera-Orefice, Robert N Lightowlers, Zofia M A Chrzanowska-Lightowlers, Alexander Galkin, Ilka Wittig, David J Pagliarini, Robert W Taylor

26. AUGUST 2025 (STOP-FSGS)

Analysis of nanomedicine primary tumor vs. metastasis targeting using clinical-stage core-crosslinked polymeric micelles

Cell Rep. 2025 Aug 26. >>[Pubmed-Link](#)<<

Larissa Yokota Rizzo, Federica De Lorenzi, Saskia von Stillfried, Seyed Mohammadali Dadfar, Qizhi Hu, Sanjay Tiwari, Claus-Christian Glüer, Marielle Thewissen, Felix Gremse, Stephan Rütten, Peter Boor, Wilhelm Jahnen-Dechent, Ruth Knüchel-Clarke, Fabian Kiessling, Cristianne J F Rijcken, Alexandros Marios Sofias, Twan Lammers

26. AUGUST 2025 (ADDRESS)

Long-term outcomes of patients with refractory cytopenia of childhood under observation only

Blood Adv. 2025 Aug 26;9(16):4279-4285. >>[Pubmed-Link](#)<<

Beatrice Drexler, Stephan Schwarz-Furlan, Irith Baumann, Martina Rudelius, Peter Nöllke, Dirk Lebrecht, Senthilkumar Ramamoorthy, Natalia Rotari, Axel Karow, Shinsuke Hirabayashi, Fabian Beier, Yvonne Lisa Behrens, Gudrun Göhring, Reinhard Kalb, Marcin W Wlodarski, Brigitte Strahm, Miriam Erlacher, Charlotte M Niemeyer, Ayami Yoshimi

26. AUGUST 2025 (MYPRED)

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Blood Adv. 2025 Aug 26;9(16):4279-4285. >>[Pubmed-Link](#)<<

Beatrice Drexler, Stephan Schwarz-Furlan, Irith Baumann, Martina Rudelius, Peter Nöllke, Dirk Lebrecht, Senthilkumar Ramamoorthy, Natalia Rotari, Axel Karow, Shinsuke Hirabayashi, Fabian Beier, Yvonne Lisa Behrens, Gudrun Göhring, Reinhard Kalb, Marcin W Wlodarski, Brigitte Strahm, Miriam Erlacher, Charlotte M Niemeyer, Ayami Yoshimi

22. AUGUST 2025 (CONNECT-GENERATE)

Obsessive-compulsive disorder – A state-of-the-art review

Neurosci Biobehav Rev. 2025 Oct. Epub 2025 Aug 22. >>[Pubmed-Link](#)<<

Dominique Endres, Miriam A Schiele, Katharina von Zedtwitz, Raphael J Dressle, Alexander Maier, Fritz Hohagen, Juan C Baldermann, Volker A Coenen, Lena Jelinek, Katharina Domschke, Ulrich Voderholzer

20. AUGUST 2025 (NEOCYST)

Systematic review of outcomes reported in clinical research on nephronophthisis: how do they align with SONG Kids priorities?

Pediatr Nephrol. 2025 Aug 20. >>[Pubmed-Link](#)<<

Mareike Dahmer-Heath, Sven Optenhövel, Tanja Hechler, Martin Konrad, Jens König

20. AUGUST 2025 (TREATHSP.NET)

Spastic Ataxia Composite (SPAXCOM): A Scale to Evaluate the Progression of Subjects with Spasticity and Ataxia

Mov Disord. 2025 Aug 20. >>[Pubmed-Link](#)<<

Cécile Di Folco, Charlotte Dubec-Fleury, Andreas Träschütz, Christoph Kessler, Selina Reich, Cynthia Gagnon, Isabelle Lessard, Xavier Rodrigue, Sirio Coccozza, Sara Satolli, Filippo M Santorelli, Alexandra Durr, Anna Heinzmann, Bart P van de Warrenburg, Ilse H J Willemse, A Nazli Başak, Atay Vural, Bernard Brais, Stephan Klebe, Rita Horvath; PROSPAX Consortium; Rebecca Schüle, Matthias Synofzik, Sophie Tezenas du Montcel

19. AUGUST 2025 (MYPRED)

PUMA-induced apoptosis drives bone marrow failure and genomic instability in telomerase-deficient mice

Cell Death Differ. 2026 Jan. Epub 2025 Aug 19. >>[Pubmed-Link](#)<<

Christian Molnar, Jovana Rajak, Julia Miriam Weiss, Irene Gonzalez-Menendez, Geoffroy Andrieux, Franziska Schreiber, Eva-Maria Kornemann, Lena Wendeburg, Gudrun Göhring, Brigitte Strahm, Fabian Beier, Doris Steinemann, Melanie Börries, Martina Rudelius, Leticia Quintanilla-Martinez, Charlotte M Niemeyer, Marena R Niewisch, Verena Labi, Sheila Bohler, Miriam Erlacher

11. AUGUST 2025 (HICHOL)

PAM-interacting domain turn-helix 51 motifs can improve Cas9-SpRY activity

Nucleic Acids Res. 2025 Aug 11. >>[Pubmed-Link](#)<<

Reto Eggenschwiler, Thomas Hoffmann, Oleg Dmytrenko, Mika Opitz, Marlene Ackel-Zakour, Pascal Wang, Shannon A McCallan, Mariane Fráguas-Eggenschwiler, Heiner Niemann, Atanas Patronov, Chase L Beisel, Tobias Cantz

11. AUGUST 2025 (CONNECT-GENERATE)

Specific viral antibodies associate with anti-NMDAR encephalitis after herpes simplex encephalitis

Brain Behav Immun. 2025 Aug 11. >>[Pubmed-Link](#)<<

Jakob Kreye, William R Morgenlander, Manjusha Thakar, Poul M Schulte-Frankenfeld, Sarah Schott, Isabel Bünger, Hans-Christian Kornau, Julia W Angkeow, Sahana Jayaraman, Carolin Otto, Wiebke Hahn, Jan Lewerenz, Franziska S Thaler, Mirjam Korporal-Kuhnke, Nico Melzer, Justina Dargvainiene, Christian G Bien, Rose Kohlie, Erik Lattwein, Dietmar Schmitz, Peter A Calabresi, Carlos A Pardo, Harald Prüss, Klemens Ruprecht, H Benjamin Larman

9. AUGUST 2025 (CONNECT-GENERATE)

Altered putamen connectivity in patients with neurological post-COVID condition

Brain Commun. 2025 Aug 9. >>[Pubmed-Link](#)<<

Lars S Schlenker, Tim J Hartung, Pia Klabunn, Katia Schwichtenberg, Josephine Heine, Lucas Adam, Christiana Franke, Carsten Finke

4. AUGUST 2025 (CONNECT-GENERATE)

Early Treatment With Intravenous Immunoglobulins and Outcomes of Patients With Anti-IgLON5 Disease

JAMA Neurol. 2025 Oct 1. Epub 2025 Aug 4. >>[Pubmed-Link](#)<<

Thomas Grüter, Carles Gaig, Yvette S Crijnen, Maarten J Titulaer, Lidia Sabater, Anna Heidebreder, Justina Dargvainiene, Anja Tietz, Stjepana Kovac, Andre Dik, María Elena Erro, Jan Lewerenz, Andrea Kraft, Frank Seifert, Romana Höftberger, Franziska S Thaler, Lucie de Azevedo, Jonathan Wickel, Juna M de Vries, Agnita J W Boon, Robin W van Steenhoven, Ralf Gold, Klaus-Peter Wandinger, Gregor Kuhlenbäumer, Josep O Dalmau, Frank Leypoldt, Francesc Graus, Ilya Ayzenberg; Anti-IgLON5 Disease Study Group

1. AUGUST 2025 (STOP-FSGS)

Non-invasive Diagnosis of Antinephrin-Associated Podocytopathy

Kidney Int Rep. 2025 May 12. eCollection 2025 Aug. >>[Pubmed-Link](#)<<

Felicitas E Hengel, Michelle C Z Chong, Wing Yin Leung, Silke Dehde, Anne Mühlig, Matthias Janneck, Henry H L Wu, Tobias B Huber, Arvind Ponnusamy, Nicola M Tomas

1. AUGUST 2025 (CONNECT-GENERATE)

Caregivers' perspectives on long-term cognitive, psychiatric, psychological, and social outcomes in NMDAR encephalitis: Initial findings from the multi-center prospective SAPIENCE study

Adv Clin Exp Med. 2025 Aug. >>[Pubmed-Link](#)<<

Marta Duda-Sikuła, Ole Jonas Boeken, Víctor Patricio, Chloe Buttard, Marie Benaiteau, Alvaro Mendes, Jerome Honnorat, Ava Easton, Josep Dalmau, Carsten Finke, Donata Kurpas

1. AUGUST 2025 (CONNECT-GENERATE)

Frequency of autoimmune-associated exogenous psychoses in routine clinical care

Brain Behav Immun Health. 2025 Jun 16. eCollection 2025 Aug. >>[Pubmed-Link](#)<<

Maria Buthut, David Haslacher, Surjo R Soekadar, Felix Machleid, Jakob Kaminski, Philipp Reber, Johanna Schoener, Anna Pichler, Moritz Thiele, Jochen Michely, Helle Foverskov-Rasmussen, Irina Baskow, Verena Rösgen-Petzold, Harald Prüss, Matthias Endres, Lasse Brandt, Andreas Heinz

1. AUGUST 2025 (STOP-FSGS)

Pathophysiological response in trauma-related acute kidney injury after blunt thoracic trauma and hemorrhagic shock in male mice

Am J Physiol Renal Physiol. 2025 Aug 1. >>[Pubmed-Link](#)<<

Rebecca Halbgebauer, Lorena Schult, Onno Borgel, Arne Maes, Florian Weißhaupt, Christina Rastner, Alitsia Ast, Ludmila Lupu, Annette Palmer, Ulrich Wachter, Stefan A Schmidt, Peter Boor, Reinhild Rösler, Sebastian Wiese, Greet Kerckhofs, Markus S Huber-Lang

30. JULI 2025 (CONNECT-GENERATE)

Reduced brain volumes in children with radiologically isolated syndrome

Mult Scler. 2025 Jul 30. >>[Pubmed-Link](#)<<

Georgia Koukou, Frederik Bartels, Annette Aigner, Robert Cleaveland, Anastasia Tuncer, Eva-Maria Wendel, Annikki Bertolini, Evangeline Wassmer, Barbara Kornek, Mareike Schimmel, Andreas Panzer, Carsten Finke, Kevin Rostásy

29. JULI 2025 (STOP-FSGS)

Kidney morphological changes associated with early-life carbonaceous ultrafine particle exposure: A pathomics approach

Environ Res. 2025 Jul 29. >>[Pubmed-Link](#)<<

Leen Rasking, Kenneth Vanbrabant, Peiqi Sun, Maartje Vangeneugden, Martin Strauch, Roel P F Schins, Flemming R Cassee, Hannelore Bové, Katrien De Vusser, Tim S Nawrot, Alessandra Tammaro, Peter Boor, Michelle Plusquin

19. JULI 2025 (CONNECT-GENERATE)

Social anxiety disorder and antibodies against glial cells in the cerebrospinal fluid

Brain Behav Immun Health. 2025 Jul 19. >>[Pubmed-Link](#)<<

Judith Weiser, Alexander Rau, Katharina von Zedtwitz, Bernd Feige, Kathrin Nickel, Simon J Maier, Raphael J Dressle, Nils Venhoff, Ludger Tebartz van Elst, Miriam A Schiele, Katharina Domschke, Harald Prüss, Dominique Endres

18. JULI 2025 (STOP-FSGS)

Pathology-oriented multiplexing enables integrative disease mapping

Nature. 2025 Jul 18. >>[Pubmed-Link](#)<<

Malte Kuehl, Yusuke Okabayashi, Milagros N Wong, Lukas Gernhold, Gabriele Gut, Nico Kaiser, Maria Schwerk, Stefanie K Gräfe, Frank Y Ma, Jovan Tanevski, Philipp S L Schäfer, Sam Mezher, Jacobo Sarabia Del Castillo, Thiago Goldbeck-Strieder, Olga Zolotareva, Michael Hartung, Fernando M Delgado Chaves, Lukas Klinkert, Ann-Christin Gnirck, Marc Spehr, David Fleck, Mehdi Joodaki, Victor Parra, Mina Shaigan, Martin Diebold, Marco Prinz, Jennifer Kranz, Johan M Kux, Fabian Braun, Oliver Kretz, Hui Wu, Florian Grahammer, Sven Heins, Marina Zimmermann, Fabian Haas, Dominik Kyllies, Nicola Wanner, Jan Czogalla, Bernhard Dumoulin, Nikolay Zolotarev, Maja Lindenmeyer, Pall Karlson, Jens R Nyengaard, Marcial Sebode, Sören Weidemann, Thorsten Wiech, Hermann-Josef Groene, Nicola M Tomas, Catherine Meyer-Schwesinger, Christoph Kuppe, Rafael Kramann, Alexandre Karras, Patrick Bruneval, Pierre-Louis Tharaux, Diego Pastene, Benito Yard, Jennifer A Schaub, Phillip J McCown, Laura Pyle, Ye Ji Choi, Takashi Yokoo, Jan Baumbach, Pablo J Sáez, Ivan Costa, Jan-Eric Turner, Jeffrey B Hodgkin, Julio Saez-Rodriguez, Tobias B Huber, Petter Bjornstad, Matthias Kretzler, Olivia Lenoir, David J Nikolic-Paterson, Lucas Pelkmans, Stefan Bonn, Victor G Puelles

12. JULI 2025 (ADDRESS)

BRCA2 Pre-mRNA Differential 5' Splicing: A Rescue of Functional Protein Properties from Pathogenic Gene Variants and a Lifeline for Fanconi Anemia D1 Patients

Int J Mol Sci. 2025 Jul 12. >>[Pubmed-Link](#)<<

Roberto Paredes, Kiran Batta, Daniel H Wiseman, Reham Gothbi, Vineet Dalal, Christine K Schmidt, Reinhard Kalb, Stefan Meyer, Detlev Schindler

7. JULI 2025 (CONNECT-GENERATE)

Comorbidities and Their Influence on Outcomes and Infectious Complications in Autoimmune Encephalitis: A Multicenter Cohort Study

Neurol Neuroimmunol Neuroinflamm. 2025 Sep. Epub 2025 Jul 7. >>[Pubmed-Link](#)<<

Amelie Bohn, Klemens Angstwurm, Christian G Bien, Kathrin Doppler, Lena Ehmke, Joachim Havla, Frank Hoffmann, Dominica Hudusch, Jaqueline Klausewitz, Franz Felix Konen, Mirjam Korporal-Kuhnke, Andrea Kraft, Tania Kümpfel, Frank Leypoldt, Marie Madlener, Lena K Pfeffer, Steffen Pfeuffer, Duygu Pul, Anna Rada, Sebastian Rauer, Christopher Sängler, Thomas Seifert-Held, Kurt-Wolfram Sühs, Franziska S Thaler, Thanos Tsaktanis, Benjamin Vlad, Klaus-Peter Wandinger, Jonathan Wickel, Simone C Tauber; GENERATE study group

7. JULI 2025 (CONNECT-GENERATE)

Intraperitoneal administration of NMDA-Subunit NR1-receptor antibodies does not improve long-term outcome in a murine MCAo-stroke model

Front Neurosci. 2025 Jul 7. >>[Pubmed-Link](#)<<

Carolin Albrecht, Christoph Harms, Jakob Kreye, Matthias Endres, Susanne Mueller, Philipp Boehm-Sturm, Stefan Paul Koch, Dorette Freyer, Harald Prüss, Samuel Knauss

7. JULI 2025 (MITONET)

Paediatric acute liver failure: A prospective, nationwide, population-based surveillance study in Germany

J Pediatr Gastroenterol Nutr. 2025 Jul 7 >>[Pubmed-Link](#)<<

Dominic Lenz, Muhammad Abdulaziz, Bianca Peters, Matias Wagner, Lea D Schlieben, Victor M Corman, Ulrich Baumann, Philip Bufler, Tal Dattner, Rainer Ganschow, Kristin Genzel, Nicole Hammann, Steffen Hartleif, Bianca Hegen, Stephan Henning, André Hoerning, Martin Jankofsky, Norman Junge, Simone Kathemann, Birgit Knoppke, Martina Kohl-Sobania, Martin Laass, Elke Lainka, Eberhard Lurz, Michael Melter, Hanna Müller, Denisa Pilic, Markus Ries, Lisa Schiefele, Tobias Schwerd, Ekkehard Sturm, Mechtild Wegner, Michael S Urschitz, Sven F Garbade, Daniel Wenning, Christian Drost, Alexander Fichtner, Stefan Kölker, Georg F Hoffmann, Holger Prokisch, Christian Staufner

3. JULI 2025 (MITONET)

Bi-allelic mutations in FASTKD5 are associated with cytochrome c oxidase deficiency and early- to late-onset Leigh syndrome

Am J Hum Genet. 2025 Jul 3. >>[Pubmed-Link](#)<<

Hana Antonicka, Woranontee Weraarpachai, Katherine M Szigety, Robert Kopajtich, James B Gibson, Johan L K Van Hove, Marisa W Friederich, Piervito Lopriore, Christiane Neuhofer, Roxanne A Van Hove, Michel A Cole, Richard Reisdorph, James T Peterson, Katherine J Dempsey, Rebecca D Ganetzky, Michelangelo Mancuso, Holger Prokisch, Eric A Shoubridge

3. JULI 2025 (STOP-FSGS)

Biofabrication of a Filtration Barrier by Integrating Electrospun Membranes and Flow in a Glomerular Co-Culture

Adv Healthc Mater. 2025 Jul 3. >>[Pubmed-Link](#)<<

Camilla Mussoni, Anna Rederer, Vladimir Stepanenko, Frank Würthner, Philipp Stahlhut, Jürgen Groll, Mario Schiffer, Taufiq Ahmad, Janina Müller-Deile

2. JULI 2025 (CONNECT-GENERATE)

Pathogenic mechanisms of autoantibodies in neurological autoimmune diseases

Curr Opin Immunol. 2025 Aug. >>[Pubmed-Link](#)<<

Siyuan Fan, Harald Prüss

2. JULI 2025 (STOP-FSGS)

Expansion and fluctuations-enhanced microscopy for nanoscale molecular profiling of cells and tissues

Nat Protoc. 2025 Jul 2. >>[Pubmed-Link](#)<<

Dominik Kyles, Hannah S Heil, Arturo G Vesga, Mario Del Rosario, Maria Schwerk, Malte Kuehl, Milagros N Wong, Victor G Puelles, Ricardo Henriques

1. JULI 2025 (TREATHSP.NET)

Long-read RNA-sequencing reveals transcript-specific regulation in human-derived cortical neurons

Open Biol. 2025 Jul. >>[Pubmed-Link](#)<<

Jishu Xu, Michaela Hörner, Elena Buena Atienza, Kalaivani Manibarathi, Maike Nagel, Stefan Hauser, Jakob Admard, Nicolas Casadei, Stephan Ossowski, Rebecca Schuele

1. JULI 2025 (CONNECT-GENERATE)

Tailored antisense oligonucleotides for ultrarare CNS diseases: An experience-based best practice framework for individual patient evaluation

Mol Ther Nucleic Acids. 2025 Jul 1. >>[Pubmed-Link](#)<<

Rebecca Schüle, Holm Graessner, Annemieke Aartsma-Rus, Willeke M C van Roon-Mom; N=1 Collaborative (N1C); 1 Mutation 1 Medicine Consortium (1M1M); Matthias Synofzik

1. JULI 2025 (CONNECT-GENERATE)

Neuropathic Pain and Distinct CASPR2 Autoantibody IgG Subclasses Drive Neuronal Hyperexcitability

Neurol Neuroimmunol Neuroinflamm. 2025 Jul. >>[Pubmed-Link](#)<<

Margarita Habib, Anna-Lena Wiessler, Patrik Fischer, Michele Niesner, Mareike Selcho, Ligia Abrante, Christian Werner, Annemarie Sodmann, Maximilian Koch, Abdolhossein Zare, Harald Prüss, Justina Dargvainiene, Jan Lewerenz, Robert Handreka, Peter Körtvelyessy, Dirk Reinhold, Franziska S Thaler, Kalliopi Pitarokoili, Robert J Kittel, Michael Briesse, Michael Sendtner, Heike Rittner, Frank Leypoldt, Claudia Sommer, Robert Blum, Kathrin Doppler, Carmen Villmann

1. JULI 2025 (STOP-FSGS)

The multifaceted role of the macrophage migration inhibitory factor family in organ fibrosis

Am J Physiol Cell Physiol. 2025 Jul 1. >>[Pubmed-Link](#)<<

Lea Herkens, Patrick Droste, Peter Boor

30. JUNI 2025 (HICHOL)

Short-term and long-term development of gut microbiota in children after liver transplantation-A prospective observational trial

Liver Transpl. 2025 Jun 30. >>[Pubmed-Link](#)<<

Imeke Goldschmidt, Miriam Kramer, Norman Junge, Niyade Ouro-Djobo, Alina Poets, Margarete Rathert, Robert Geffers, Ulrich Baumann, Björn Hartleben, Kora Dorothea Schulze, Sabrina Woltemate, Marius Vital

25. JUNI 2025 (STOP-FSGS)

The differential expression of MAGI2 in glomerulopathies and its application as a molecular discriminator of podocytopathies

J Transl Med. 2025 Jun 25. >>[Pubmed-Link](#)<<

Florian Siegerist, Eleonora Hay, Elke Hammer, Anna Iervolino, Claudia Weber, Juan Saydou Dikou, Eleni Stamellou, Linus Butt, Thomas Benzing, Thorsten Wiech, Paul T Brinkötter, Uwe Zimmerman, Giovambattista Capasso, Christos Chatziantoniou, Christos E Chadichristos, Tobias B Huber, Uwe Völker, Maximilian Schindler, Nicole Endlich

23. JUNI 2025 (STOP-FSGS)

Loss of genome maintenance is linked to mTOR complex 1 signaling and accelerates podocyte damage

JCI Insight. 2025 May 20. eCollection 2025 Jun 23. >>[Pubmed-Link](#)<<

Fabian Braun, Amrei M Mandel, Linda Blomberg, Milagros N Wong, Georgia Chatzinikolaou, David H Meyer, Anna Reinelt, Viji Nair, Roman Akbar-Haase, Phillip J McCown, Fabian Haas, He Chen, Mahdieh Rahmatollahi, Damian Fermin, Robin Ebbestad, Gisela G Slaats, Tillmann Bork, Christoph Schell, Sybille Koehler, Paul T Brinkkoetter, Maja T Lindenmeyer, Clemens D Cohen, Martin Kann, David Unnersjö-Jess, Wilhelm Bloch, Matthew G Sampson, Martijn Et Dollé, Victor G Puellas, Matthias Kretzler, George A Garinis, Tobias B Huber, Bernhard Schermer, Thomas Benzing, Björn Schumacher, Christine E Kurschat

10. JUNI 2025 (MYPRED)

Hematological phenotypes in GATA2 deficiency syndrome arise from aging, maladaptation to proliferation, and somatic events

Blood Adv. 2025 Jun 10. >>[Pubmed-Link](#)<<

Juncal Fernandez-Orth, Cansu Koyunlar, Julia M Weiss, Emanuele Gioacchino, Hans de Looper, Geoffroy Andrieux, Mariëtte Ter Borg, Joke Zink, Irene Gonzalez-Menendez, Remco Hoogenboezem, Baris Yigit, Kirsten J Gussinklo, Roger Mulet-Lazaro, Charlotte Wantzen, Sophie Pfeiffer, Christian Molnar, Eric Bindels, Sheila Bohler, Mathijs Sanders, Leticia Quintanilla-Martinez, Marcin Wlodarski, Melanie Boerries, Ivo P Touw, Charlotte Niemeyer, Miriam Erlacher, Emma de Pater

6. JUNI 2025 (MYPRED)

Biallelic SH2B3 germline variants are associated with a neonatal myeloproliferative disease and multisystemic involvement

Eur J Hum Genet. 2025 Jun 6. >>[Pubmed-Link](#)<<

Davide Leardini, Elisabetta Flex, Elliot Stieglitz, Sara Cerasi, Salvatore Nicola Bertuccio, Francesco Baccelli, Krisztián Kállay, Paula Kjollerstrom, Sara Batalha, Giovanna Carpentieri, Lucia Pedace, Andrea Cioffi, Mahmoud Hammad, Maria Miranda, Marta Rojas, Anupama Rao, Andrew J Innes, Martina Rudelius, Valeria Santini, Marco Raddi, Kok-Hoi Teh, Rita De Vito, Ayami Yoshimi, Marco Tartaglia, Franco Locatelli, Charlotte M Niemeyer, Riccardo Masetti

5. JUNI 2025 (TREATHSP.NET)

Outcome measures of instrumented gait analysis in hereditary spastic paraplegia: a systematic review

J Neuroeng Rehabil. 2025 Jun 5. >>[Pubmed-Link](#)<<

Veronika Koch, Alzhaa Ibrahim, Juergen Winkler, Bjoern Eskofier, Martin Regensburger, Heiko Gassner

5. JUNI 2025 (TREAT-ION)

The genetic and phenotypic spectrum of GABRB1-related disorders

Brain. 2025 Jun 5. >>[Pubmed-Link](#)<<

Charissa Millevirt, Anthony Sze Hon Kan, Moritz Hanke, Mahmoud Koko, Maryam Erfanian Omidvar, Ulrike B S Hedrich, Thomas V Wuttke, Nina Barišić, Lieven Lagae, Ángel Aledo-Serrano, Eva-Maria Niehoff, Konrad Platzer, Pia Zacher, Tilman Polster, Robertino Dilella, Edoardo Monfrini, David Geneviève, Agathe Roubertie, Ange-Line Bruel, Frederic Tran Mau-Them, Majed Dasouki, Stacey Cohen, Ingo Helbig, Alicia G Harrison, Ellis Colin, Holly A Dubbs, Eric D Marsh, Sébastien Lebon, Na He, Heng Meng, Mary Chebib, Rikke S Møller, Carla Marini, Philip K Ahring, Holger Lerche, Sarah Weckhuysen

5. JUNI 2025 (GAIN)

Structural basis for OAS2 regulation and its antiviral function

Mol Cell. 2025 Jun 5 >>[Pubmed-Link](#)<<

Veronika Merold, Indra Bekere, Stefanie Kretschmer, Adrian F Schnell, Dorota Kmiec, Rinu Sivarajan, Katja Lammens, Rou Liu, Julia Mergner, Julia Teppert, Maximilian Hirschenberger, Alexander Henrici, Sarah Hammes, Kathrin Bude, Marcus Weitz, Karl Hackmann, Lars M Koenig, Andreas Pichlmair, Nadine Schwierz, Konstantin M J Sparrer, Min Ae Lee-Kirsch, Carina C de Oliveira Mann

4. JUNI 2025 (MYPRED)

Flavopiridol restores granulopoiesis in experimental models of severe congenital neutropenia

Mol Ther. 2025 Jun 4. >>[Pubmed-Link](#)<<

Masoud Nasri, Benjamin Dannenmann, Larissa Doll, Betül Findik, Franka Bernhard, Sergey Kandabarau, Maksim Klimiankou, Meinrad Gawaz, Claudia Lengerke, Cornelia Zeidler, Karl Welte, Julia Skokowa

4. JUNI 2025 (MITONET)

Autosomal Recessive Cerebellar Ataxias: Translating Genes to Therapies

Ann Neurol. 2025 Jun 4. >>[Pubmed-Link](#)<<

Brent L Fogel, Thomas Klopstock, David R Lynch, Francesca Maltecca, Mayank Verma, Berge A Minassian, Frances M Platt, Débora Farina Gonçalves, Hélène Puccio, Andreas Roos, Matthis Synofzik

4. JUNI 2025 (CONNECT-GENERATE)

Association of ADHD symptoms, pain, and tics with anti-thalamus antibodies in cerebrospinal fluid

J Neuroimmunol. 2025 Sep 15. Epub 2025 Jun 4. >>[Pubmed-Link](#)<<

Dominique Endres, Katharina von Zedtwitz, Alexander Rau, Bernd Feige, Hansjörg Mast, Alexander Maier, Marco Reisert, Kathrin Nickel, Joachim Brumberg, Tobias Boettler, Cornelia Glaser, Nils Venhoff, Horst Urbach, Juan C Baldermann, Katharina Domschke, Ludger Tebartz van Elst, Luciana Hannibal, Harald Prüss, Simon J Maier

1. JUNI 2025 (GAIN)

Clinical manifestations, disease penetrance, and treatment in individuals with SOCS1 insufficiency: a registry-based and population-based study

Lancet Rheumatol. 2025 Jun. >>[Pubmed-Link](#)<<

Jerome Hadjadj, Anna Wolfers, Oleg Borisov, Derek Hazard, Ronan Leahy, Marie Jeanpierre, Alexandre Belot, Shahrzad Bakhtiar, Fabian Hauck, Pui Y Lee, Stefano Volpi, Serena Palmeri, Vincent Barlogis, Nathalie Aladjidi, Georg Ebetsberger-Dachs, Jerome Avouac, Fabienne Charbit-Henrion, Morgane Cheminant, Jean Donadieu, Sujal Ghosh, David P Hoytema van Konijnenburg, Julia Körholz, Jacinta Bustamante, Jeremie Rosain, Lisa Forbes Satter, Ingrid Selmerud, Georgios Sogkas, Benedicte Neven, Frederic Rieux-Laucat, Stephan Ehl; SOCS1 Study Group

1. JUNI 2025 (TREATHSP.NET)

Unravelling axonal transcriptional landscapes: insights from induced pluripotent stem cell-derived cortical neurons and implications for motor neuron degeneration

Open Biol. 2025 Jun. >>[Pubmed-Link](#)<<

Jishu Xu, Michaela Hörner, Maike Nagel, Perwin Perhat, Milena Korneck, Marvin Noß, Stefan Hauser, Ludger Schoels, Jakob Admard, Nicolas Casadei, Rebecca Schuele

27. MAI 2025 (GAIN)

Expansion of CD57+ CD8 T cells in common variable immunodeficiency with hepatopathy and CMV infection

Front Immunol. 2025 May 27. >>[Pubmed-Link](#)<<

Patrick Bez, Enrico Santangeli, Sigune Goldacker, Ulrich Salzer, Klaus Warnatz

22. MAI 2025 (TREATHSP.NET)

Microglia activation orchestrates CXCL10-mediated CD8+ T cell recruitment to promote aging-related white matter degeneration

Nat Neurosci. 2025 May 22. >>[Pubmed-Link](#)<<

Janos Groh, Ruqing Feng, Xidi Yuan, Lu Liu, Dennis Klein, Gladis Hutahaeen, Elisabeth Butz, Zhen Wang, Lisa Steinbrecher, Jonas Neher, Rudolf Martini, Mikael Simons

16. MAI 2025 (CONNECT-GENERATE)

MOG-Encephalitis is the Most Prevalent Autoimmune Encephalitis in Children: MERIN Study Data on Encephalitis

Neuropediatrics. 2025 May 16. >>[Pubmed-Link](#)<<

Ruth Helena Fellmeth, Lampros Kousoulos, George Christoph Korenke, Hans-Jürgen Christen, Masyar Monazahian, Justine Dargvainiene, Klaus-Peter Wandinger, Frank Leyboldt, Kevin Rostásy

16. MAI 2025 (TREATHSP.NET)

RNA splicing modulator for Huntington's disease treatment induces peripheral neuropathy

iScience. 2025 Apr 8. eCollection 2025 May 16. >>[Pubmed-Link](#)<<

Florian Krach, Tom Boerstler, Stephanie Neubert, Laura Krumm, Martin Regensburger, Jürgen Winkler, Beate Winner

1. MAI 2025 (MITONET)

280th ENMC International Workshop: The ERN EURO-NMD mitochondrial diseases workinggroup; diagnostic criteria and outcome measures in primary mitochondrial myopathies. Hoofddorp, the Netherlands, 22-24 November 2024

Neuromuscul Disord. 2025 May. >>[Pubmed-Link](#)<<

Michelangelo Mancuso, Piervito Lopriore, Luisa Semmler, Cornelia Kornblum; 280th ENMC workshop study group

1. MAI 2025 (GAIN)

Uncovering risk factors of premature mortality in Common Variable Immunodeficiency (CVID)

J Allergy Clin Immunol Pract. 2025 May. >>[Pubmed-Link](#)<<

Patrick Bez, Bas Smits, Christoph Geier, Aleksandra Hirsch, Andrés Caballero de Oyteza, Michele Proietti, Bodo Grimbacher, Martin Wolkewitz, Sigune Goldacker, Klaus Warnatz

1. MAI 2025 (CONNECT-GENERATE)

Autoimmune-associated epilepsy – a challenging concept

Seizure. 2025 May. >>[Pubmed-Link](#)<<

Nico Melzer, Felix Rosenow

1. MAI 2025 (STOP-FSGS)

Newer B-cell and plasma-cell targeted treatments for rituximab-resistant patients with membranous nephropathy

Clin Kidney J. 2025 Mar 21. eCollection 2025 May. >>[Pubmed-Link](#)<<

Marc Buse, Evangelia Dounousi, Rafael Kramann, Jürgen Floege, Eleni Stamellou

1. MAI 2025 (NEOCYST)

A risk score to predict kidney survival in patients with autosomal recessive polycystic kidney disease at the age of two months

Kidney Int. 2025 May. >>[Pubmed-Link](#)<<

Kathrin Burgmaier, Samuel Kilian, Klaus Arbeiter, Bahriye Atmis, Olivia Boyer, Anja Buescher, Ismail Dursun, Florian Erger, Marc Fila, Matthias Galiano, Ibrahim Gokce, Karsten Haeffner, Dieter Haffner, Nakysa Hooman, Guenter Klaus, Jens König, Bärbel Lange-Sperandio, Matko Marlais, Laura Massella, Djalila Mekahli, Monika Miklaszewska, Gordana Miloševski-Lomić, Lukasz Obrycki, Bruno Ranchin, Barbara Seitz, Stella Stabouli, Yilmaz Tabel, Katarzyna Taranta-Janusz, Lutz Thorsten Weber, Marcus Weitz, Elke Wühl, Alev Yilmaz, Jörg Dötsch, Franz Schaefer, Max Christoph Liebau; ARegPKD consortium

30. APRIL 2025 (MITONET)

Pleiotropic effects of MORC2 derive from its epigenetic signature

Brain. 2026 Jan 8. Epub 2025 Apr 30. >>[Pubmed-Link](#)<<

Fatemeh Peymani, Tomohiro Ebihara, Dmitrii Smirnov, Robert Kopajtich, Masahiro Ando, Enrico Bertini, Rosalba Carrozzo, Daria Diodato, Felix Distelmaier, Fang Fang, Daniele Ghezzi, Maja Hempel, Katarzyna Iwanicka-Pronicka, Thomas Klopstock, Sarah L Stenton, Costanza Lamperti, Zhimei Liu, Aysylu Murtazina, Yuji Okamoto, Yasushi Okazaki, Dorota Piekutowska-Abramczuk, Agnès Rötig, Oxana Ryzhkova, Christian Schlein, Olga Shagina, Hiroshi Takashima, Polina G Tsygankova, Michael Zech, Thomas Meitinger, Masaru Shimura, Kei Murayama, Holger Prokisch

29. APRIL 2025 (HICHO)

Pediatric Liver and Kidney Transplant Recipients Demonstrate Greater Serological Response to SARS-CoV-2 Vaccination Than Adults

Transplant Direct. 2025 Apr 29. eCollection 2025 May. >>[Pubmed-Link](#)<<

Tobias Laue, Maria Pilar Ballester, Lily Meoli, Carl Grabitz, Eva Uson, Lorenzo D Antiga, Valerie McLin, Montserrat Pujadas, Ângela Carvalho-Gomes, Ivan Sahuco, Ariadna Bono, Federico D'Amico, Raffaella Viganò, Elena Diago, Beatriz Tormo Lanseros, Elvira Inglese, Dani Martinez Vazquez, Annelotte Broekhoven, Marjolein Kikkert, Shessy P Torres Morales, Sebenzile K Myeni, Mar Riveiro-Barciela, Adriana Palom, Nicola Zeni, Alessandra Brocca, Annarosa Cussigh, Sara Cmet, Maria Desamparados Escudero-García, Matteo Stocco, Leonardo Antonio Natola, Donatella Ieluzzi, Veronica Paon, Angelo Sangiovanni, Elisa Farina, Clara Dibenedetto, Yolanda Sánchez-Torrijos, Ana Lucena-Varela, Eva Román, Elisabet Sánchez, Rubén Sánchez-Aldehuelo, Julia López-Cardona, Dhaarica Jeyanesan, Alejandro Esquivel Morocho, Itzel Canas-Perez, Christine Eastgate, Simone Di Cola, Lucia Lapenna, Giacomo Zaccherini, Deborah Bongiovanni, Antonio Riva, Rajni Sharma, Hio Lam Phoebe Tsou, Nicola Harris, Paola Zanaga, Katia Sayaf, Sabir Hossain, Javier Crespo, Mercedes Robles-Díaz, Antonio Madejón, Helena Degroote, Marko Korenjak, Xavier Verhelst, Javier García-Samaniego, Raúl J Andrade, Paula Iruzubieta, Gavin Wright, Paolo Caraceni, Manuela Merli, Vishal C Patel, Amir Gander, Agustín Albillos, Germán Soriano, Maria Francesca Donato, David Sacerdoti, Pierluigi Toniutto, Maria Buti, Christophe Duvoux, Paolo Antonio Grossi, Thomas Berg, Wojciech G Polak, Massimo Puoti, Anna Bosch-Comas, Luca S Belli, Patrizia Burra, Francesco Paolo Russo, Minneke Coenraad, José Luis Calleja, Giovanni Perricone, Shilpa Chokshi, Marina Berenguer, Joan Clària, Richard Moreau, Javier Fernández, Vicente Arroyo, Paolo Angeli, Cristina Sánchez-Garrido, Javier Ampuero, Salvatore Piano, Emanuele Nicastro, Nathalie Rock, Debbie Shawcross, Lindsey Edwards, Frauke Mutschler, Anette Melk, Gautam Mehta, Ulrich Baumann, Rajiv Jalan

25. APRIL 2025 (STOP-FSGS)

Nanoscale imaging of biological systems via expansion and super-resolution microscopy

Appl Phys Rev. 2025 April 25 2025. >>[AIP-Link](#)<<

Daria Aristova, Dominik Kyllies, Mario Del Rosario, Hannah S. Heil, Maria Schwerk, Malte Kuehl, Milagros N., Ricardo Henriques, Victor G. Puelles

25. APRIL 2025 (MITONET)

Expanding the Allelic and Clinical Heterogeneity of Movement Disorders Linked to Defects of Mitochondrial Adenosine Triphosphate Synthase

Mov Disord. 2025 Apr 25. >>[Pubmed-Link](#)<<

Philip Harrer, Magdalena Krygier, Martin Krenn, Volker Kittke, Martin Danis, Georgi Krastev, Alice Saporov, Virginie Pichon, Marlène Malbos, Clarisse Scherer, Ivana Dzinovic, Matej Skorvanek, Robert Kopajtich, Holger Prokisch, Sara Silvaieh, Anna Grisold, Maria Mazurkiewicz-Beldzińska, Jean-Madeleine de Sainte Agathe, Juliane Winkelmann, Jan Necpal, Robert Jech, Michael Zech

24. APRIL 2025 (CONNECT-GENERATE)

Obsessive-Compulsive Symptoms and Heterozygous Microduplication of the RB1CC1 Gene

Biol Psychiatry Glob Open Sci. 2025 Apr 24. >>[Pubmed-Link](#)<<

Alexander Maier, Katharina von Zedtwitz, Simon J Maier, Bernd Feige, Marco Reisert, Volker A Coenen, Alexander Rau, Raphael J Dressle, Kathrin Nickel, Miriam A Schiele, Birgitta Gläser, Katharina Domschke, Katalin Komlosi, Dominique Endres

22. APRIL 2025 (CONNECT-GENERATE)

From immunobiology to intervention: Pathophysiology of autoimmune encephalitis

Semin Immunol. 2025 Apr 22. >>[Pubmed-Link](#)<<

Marie A Homeyer, Alice Falck, Lucie Y Li, Harald Prüss

18. APRIL 2025 (CONNECT-GENERATE)

Lymphocyte signatures correspond to clinical phenotypes in autoimmune limbic encephalitis

Brain Commun. 2025 Apr 18. >>[Pubmed-Link](#)<<

Saskia Räuber, Andreas Schulte-Mecklenbeck, Kelvin Sarink, Christoph Müller, Manoj Mannil, Lisa Langenbruch, Andre Dik, Sumanta Barman, Christine Strippel, Marco Gallus, Kristin S Golombeck, Christina B Schroeter, Alice Willison, Christopher Nelke, Fatme Seval Ismail, Wolfram Schwindt, Norbert Goebels, Stjepana Kovac, Heinz Wiendl, Gerd Meyer Zu Hörste, Thomas Duning, Michael Hanke, Tobias Ruck, Walter Heindel, Udo Dannlowski, Tim Hahn, Catharina C Gross, Sven G Meuth, Nico Melzer

17. APRIL 2025 (NEOCYST)

Non-invasive screening for liver fibrosis by acoustic radiation force impulse in patients with ciliopathies

Sci Rep. 2025 Apr 17. >>[Pubmed-Link](#)<<

Johanna Bresch, Jens König, Martin Konrad, Sabine Kollmann, Mareike Dahmer-Heath, Hauke Sebastian Heinzow, Michael Praktijn, Jonel Trebicka, Carsten Bergmann, Hartmut H-J Schmidt, Bernhard Schlevogt

16. APRIL 2025 (STOP-FSGS)

Deep Visual Proteomics maps proteotoxicity in a genetic liver disease

Nature. 2025 Apr 16. >>[Pubmed-Link](#)<<

Florian A Rosenberger, Sophia C Mädler, Katrine Holtz Thorhauge, Sophia Steigerwald, Malin Fromme, Mikhail Lebedev, Caroline A M Weiss, Marc Oeller, Maria Wahle, Andreas Metousis, Maximilian Zwiebel, Niklas A Schmacke, Sönke Detlefsen, Peter Boor, Ondřej Fabián, Soňa Fraňková, Aleksander Krag, Pavel Strnad, Matthias Mann

15. APRIL 2025 (ADDRESS)

Genetic inactivation of FAAP100 causes Fanconi anemia due to disruption of the monoubiquitin ligase core complex

J Clin Invest. 2025 Apr 15. >>[Pubmed-Link](#)<<

Julia Kuehl, Yutong Xue, Fenghua Yuan, Ramanagouda Ramanagoudr-Bhojappa, Simone Pickel, Reinhard Kalb, Settara Chandrasekharappa, Weidong Wang, Yanbin Zhang, Detlev Schindler

15. APRIL 2025 (NEOCYST)

Urinary peptide signature distinguishes autosomal recessive polycystic kidney disease from other causes of chronic kidney disease

Clin Kidney J. 2025 Apr 15. >>[Pubmed-Link](#)<<

Kathrin Burgmaier, Bénédicte Buffin-Meyer, Julie Klein, Brian Becknell, Daryl McLeod, Jan Boeckhaus, Oliver Gross, Claudia Dafinger, Justyna Siwy, Stéphane Decramer, Franz Schaefer, Max C Liebau, Joost P Schanstra

14. APRIL 2025 (TREATHSP.NET)

Unraveling undiagnosed rare disease cases by HiFi long-read genome sequencing

Genome Res. 2025 Apr 14. >>[Pubmed-Link](#)<<

Wouter Steyaert, Lydia Sagath, German Demidov, Vicente A Yépez, Anna Esteve-Codina, Julien Gagneur, Kornelia Ellwanger, Ronny Derks, Marjan Weiss, Amber den Ouden, Simone van den Heuvel, Hilde Swinkels, Nick Zomer, Marloes Steehouwer, Luke O’Gorman, Galuh Astuti, Kornelia Neveling, Rebecca Schüle, Jishu Xu, Matthis Synofzik, Danique Beijer, Holger Hengel, Ludger Schöls, Kristl G Claeys, Jonathan Baets, Liedewei Van de Vondel, Alessandra Ferlini, Rita Selvatici, Heba Morsy, Marwa Saeed Abd Elmaksoud, Volker Straub, Juliane Müller, Veronica Pini, Luke Perry, Anna Sarkozy, Irina Zaharieva, Francesco Muntoni, Enrico Bugiardini, Kiran Polavarapu, Rita Horvath, Evan Reid, Hanns Lochmüller Marco Spinazzi, Marco Savarese; Solve-RD DITF-ITHACA, Solve-RD DITF-Euro-NMD, Solve-RD DITF-RND, Solve-RD DITF-EpiCARE; Leslie Matalonga, Steven Laurie, Han G Brunner, Holm Graessner, Sergi Beltran, Stephan Ossowski, Lisenka E L M Vissers, Christian Gilissen, Alexander Hoischen; Solve-RD consortium

11. APRIL 2025 (HICHOL)

ECM1 expression in chronic liver disease: Regulation by EGF/STAT1 and IFN γ /NRF2 signalling

JHEP Rep. 2025 Apr 11. >>[Pubmed-Link](#)<<

Yujia Li, Chenjun Huang, Weiguo Fan, Seddik Hammad, Cyrill Géraud, Lea Berger, Shanshan Wang, Ye Yao, Chenhao Tong, Claudia Rubie, Laura Kim Feiner, Zeribe C Nwosu, Frederik Link, Pia Erdösi, Weronika Piorońska, Kerry Gould, Christoph Meyer, Rilu Feng, Hui Liu, Chen Shao, Bing Sun, Huiguo Ding, Roman Liebe, Matthias P A Ebert, Honglei Weng, Matthias Glanemann, Chunfang Gao, Peter Ten Dijke, Steven Dooley, Sai Wang

1. APRIL 2025 (MITONET)

Natural History of Patients With Mitochondrial ATPase Deficiency Due to Pathogenic Variants of MT-ATP6 and MT-ATP8

Neurology. 2025 Apr. >>[Pubmed-Link](#)<<

Sara Carli, Anna Levarlet, Daria Diodato, Enrico Silvio Bertini, Diego Martinelli, Alessandro Malandrini, Diego Lopercolo, Gian Nicola Gallus, Rebecca D Ganetzky, Chiara La Morgia, Valerio Carelli, Guido Primiano, Cristina Domínguez-González, Pablo Serrano-Lorenzo, Miguel A Martín, Anna Ardisson, Costanza Lamperti, Valeria Nicoletta, Thomas Klopstock, Felix Distelmaier, Leopold Zeng, Boriana Büchner, Michelangelo Mancuso, Markus Schuelke, Alessandro Prigione, Caterina Garone

1. APRIL 2025 (TREAT-ION)

Comprehensive analysis of human dendritic spine morphology and density

J Neurophysiol. 2025 Apr 1. >>[Pubmed-Link](#)<<

Kerstin D Schünemann, Roxanne M Hattingh, Matthijs B Verhoog, Danqing Yang, Aniella V Bak, Sabrina Peter, Karen M J van Loo, Stefan Wolking, Deborah Kronenberg-Versteeg, Yvonne Weber, Niklas Schwarz, Joseph V Raimondo, Roger Melvill, Sean A Tromp, James T Butler, Anke Höllig, Daniel Delev, Thomas V Wuttke, Björn M Kampa, Henner Koch

1. APRIL 2025 (TREAT-ION)

The severity of SLC1A2-associated neurodevelopmental disorders correlates with transporter dysfunction

EBioMedicine. 2025 Apr. >>[Pubmed-Link](#)<<

Peter Kovermann, Allan Bayat, Christina D Fenger, Lisette Leeuwen, Artem Borovikov, Artem Sharkov, Virginie Levrat, Gaetan Lesca, Laurence Perrin, Jonathan Levy, Christoph Fahlke, Rikke S Møller, Anders A Jensen

24. MÄRZ 2025 (TREATHSP.NET)

Smartphone Application for Spastic Ataxias Cross-Sectional Validation of a Newly Developed Smartphone App for Remote Monitoring in Spastic Ataxias

Cerebellum. 2025 Mar 24. >>[Pubmed-Link](#)<<

Ilse H J Willemse, Sabato Mellone, Carlo Tacconi, Winfried Ilg, Rebecca Schüle, Matthis Synofzik, Jorik H Nonnekes, Bart P C van de Warrenburg

24. MÄRZ 2025 (STOP-FSGS)

A new era in nephrology: the role of super-resolution microscopy in research, medical diagnostic, and drug discovery

Kidney Int. 2025 Mar 24. >>[Pubmed-Link](#)<<

Florian Siegerist, Kirk N Campbell, Nicole Endlich

17. MÄRZ 2025 (CONNECT-GENERATE)

Activated $\alpha\beta$ T- and reduced mucosa-associated invariant T cells in LGI1- and CASPR2-encephalitis

Brain. 2025 Mar 17 (Epub ahead of print). >>[Pubmed-Link](#)<<

Daniela Esser, Louisa Müller-Miny, Michael Heming, Manuela Paunovic, Martijn van Duijn, Ligia Abrante Cabrera, Katharina M Mair, Christine Strippel, Saskia Räuber, Justina Dargvainiene, Stjepana Kovac, Catharina C Gross, Nina Fransen, Robin van Steenhoven, Péter Körtvélyessy 8, Werner Stenzel, Romana Höftberger, Eric Bindels, Christian G Bien, Heinz Wiendl, Sven G Meuth, Jan Bauer, Nico Melzer, Maarten J Titulaer, Frank Leypoldt, Gerd Meyer Zu Hörste; in cooperation with the EMC-AIE Study group

8. MÄRZ 2025 (MYPRED)

Oncogenic and microenvironmental signals drive cell type specific apoptosis resistance in juvenile myelomonocytic leukemia

Cell Death Dis. 2025 Mar 8. >>[Pubmed-Link](#)<<

Naile Koleci, Ying Wu, Niels Anton Wehner, Jovana Rajak, Venugopal Rao Mittapalli, Julia Mergner, Hui Xiao, Jun Wang, Madeleine Wahl, Sheila Bohler, Konrad Aumann, Georg Häcker, Senthilkumar Ramamoorthy, Melanie Boerries, Susanne Kirschnek, Miriam Erlacher

4. MÄRZ 2025 (CONNECT-GENERATE)

Differences in innate immune cell populations distinguish autoimmune from herpesvirus-associated encephalitis

J Autoimmun. 2025 Mar 4. >>[Pubmed-Link](#)<<

Saskia Räuber, Andreas Schulte-Mecklenbeck, Kelvin Sarink, Kristin S Golombeck, Christina B Schroeter, Alice Willison, Christopher Nelke, Christine Strippel, Andre Dik, Marco Gallus, Stjepana Kovac, Heinz Wiendl, Gerd Meyer Zu Hörste, Tobias Ruck, Oliver M Grauer, Udo Dannlowski, Tim Hahn, Catharina C Gross, Sven G Meuth, Nico Melzer

1. MÄRZ 2025 (CONNECT-GENERATE)

Cognitive Deficits in Anti-LGI1 Encephalitis Are Linked to Immunotherapy-Resistant White Matter Network Changes

Neurol Neuroimmunol Neuroinflamm. 2025 Mar. >>[Pubmed-Link](#)<<

Stephan Krohn, Leonie Müller-Jensen, Joseph Kuchling, Amy Romanello, Katharina Wurdack, Sophia Rekers, Thorsten Bartsch, Frank Leypoldt, Friedemann Paul, Christoph J Ploner, Harald Prüss, Carsten Finke

28. FEBRUAR 2025 (TREATHSP.NET)

Novel co-culture model of T cells and midbrain organoids for investigating neurodegeneration in Parkinson's disease

NPJ Parkinsons Dis. 2025 Feb 28. >>[Pubmed-Link](#)<<

Elizaveta Gerasimova, Amke C Beenen, Daniil Kachkin, Martin Regensburger, Sebastian Zundler, David B Blumenthal, Gloria Lutzny-Geier, Beate Winner, Iryna Prots

15. FEBRUAR 2025 (CONNECT-GENERATE)

Factors associated with reduction in quality of life after SARS-CoV-2 infection

Sci Rep. 2025 Feb 26;15(1):6833. >>[Pubmed-Link](#)<<

Christian Neumann, Tim J Hartung, Klara Boje, Thomas Bahmer, Julian Keil, Wolfgang Lieb, Katrin Franzpoetter, Julius Welzel, Irina Chaplinskaya-Sobol, Matthias Endres, Johanna Geritz, Karl Georg Haeusler, Peter Heuschmann, Andreas Hinz, Sina Hopff, Anna Horn, Carolin Nuernberger, Lena Schmidbauer, Michael Krawczak, Anne-Kathrin Ruß, Lilian Krist, Thomas Keil, Jennifer Kudelka, Corina Maetzler, Anja Mehnert-Theuerkauf, Felipe A Montellano, Caroline Morbach, Sein Schmidt, Jan Heyckendorf, Flo Steigerwald, Stefan Stoerk, Christina Lemhoefer, Stefan Schreiber, Carsten Finke, Walter Maetzler

12. FEBRUAR 2025 (MITONET)

Combined genomics and proteomics unveils elusive variants and vast aetiologic heterogeneity in dystonia

Brain. 2025 Feb 12. >>[Pubmed-Link](#)<<

Michael Zech, Ivana Dzinovic, Matej Skorvanek, Philip Harrer, Jan Necpal, Robert Kopajtich, Volker Kittke, Erik Tilch, Chen Zhao, Eugenia Tsoma, Ugo Sorrentino, Elisabetta Indelicato, Antonia Stehr, Alice Saparov, Lucia Abela, Miriam Adamovicova, Alexandra Afenjar, Birgit Assmann, Janette Baloghova, Matthias Baumann, Riccardo Berutti, Zuzana Brezna, Melanie Brugger, Theresa Brunet, Benjamin Cogne, Isabel Colangelo, Erin Conboy, Felix Distelmaier, Matthias Eckenweiler, Barbara Garavaglia, Arie Geerlof, Elisabeth Graf, Annette Hackenberg, Denisa Harvanova, Bernhard Haslinger, Petra Havrankova, Georg F Hoffmann, Wibke G Janzarik, Boris Keren, Miriam Kolnikova, Konstantinos Kolokotronis, Zuzana Kosutzka, Anne Koy, Martin Krenn, Magdalena Krygier, Katarina Kusikova, Oliver Maier, Thomas Meitinger, Christian Mertes, Ivan Milenkovic, Edoardo Monfrini, Andre Santos Dias Mourao, Thomas Musacchio, Mathilde Nizon, Miriam Ostrozovicova, Martin Pavlov, Iva Prihodova, Irena Rektorova, Luigi M Romito, Barbora Rybanska, Ariane Sadr-Nabavi, Susanne Schwenger, Ali Shoeibi, Alexandra Sitzberger, Dmitrii Smirnov, Jana Svantnerova, Raushana Tautanova, Sandra P Toelle, Olga Ulmanova, Francesco Vetrini, Katharina Vill, Matias Wagner, David Weise, Giovanna Zorzi, Alessio Di Fonzo, Konrad Oexle, Steffen Berweck, Volker Mall, Sylvia Boesch, Barbara Schormair, Holger Prokisch, Robert Jech, Juliane Winkelmann

12. FEBRUAR 2025 (MITONET)

Leber's hereditary optic neuropathy – current status of idebenone and gene replacement therapies

Med Genet. 2025 Feb 12. >>[Pubmed-Link](#)<<

Thomas Klopstock, Leopold H Zeng, Claudia Priglinger

12. FEBRUAR 2025 (GAIN)

CD21low B cells reveal a unique glycosylation pattern with hypersialylation and hyperfucosylation

Front Immunol. 2025 Feb 12. >>[Pubmed-Link](#)<<

Peter Tobias Felixberger, Geoffroy Andrieux, Andrea Maul-Pavicic, Sigune Goldacker, Ina Harder, Sylvia Gutenberger, Jonathan J M Landry, Vladimir Benes, Till Fabian Jakob, Melanie Boerries, Lars Nitschke, Reinhard Edmund Voll, Klaus Warnatz, Baerbel Keller

11. FEBRUAR 2025 (GAIN)

The intestinal microbiome and metabolome discern disease severity in cytotoxic T-lymphocyte-associated protein 4 deficiency

Microbiome. 2025 Feb 11. >>[Pubmed-Link](#)<<

Prabha Chandrasekaran, Máté Krausz, Yu Han, Noriko Mitsui, Annemarie Gabrysch, Christina Nöltner, Michele Proietti, Theo Heller, Caroline Grou, Virginia Calderon, Poorani Subramanian, Drew R Jones, Yik Siu, Clayton Deming, Sean Conlan, Steven M Holland, Julia A Segre, Gulbu Uzel, Bodo Grimbacher, Emilia Liana Falcone

3. FEBRUAR 2025 (TREAT-ION)

Gene-replacement therapy in neurodevelopmental disorders: progress and challenges

J Clin Invest. 2025 Feb 3. >>[Pubmed-Link](#)<<

Holger Lerche, Ulrike Bs Hedrich, Thomas V Wuttke

2. FEBRUAR 2025 (ADDRESS)

MSH2, MSH6, MLH1, and PMS2 immunohistochemistry as highly sensitive screening method for DNA mismatch repair deficiency syndromes in pediatric high-grade glioma

Acta Neuropathol. 2025 Feb 2. >>[Pubmed-Link](#)<<

Lea L Friker, Thomas Perwein, Andreas Waha, Evelyn Dörner, Rebecca Klein, Mirjam Blattner-Johnson, Julian P Layer, Dominik Sturm, Gunther Nussbaumer, Robert Kwiecien, Isabel Spier, Stefan Aretz, Kornelius Kerl, Ulrike Hennewig, Marius Rohde, Axel Karow, Ingmar Bluemcke, Ann Kristin Schmitz, Harald Reinhard, Pablo Hernáiz Driever, Susanne Wendt, Annette Weiser, Ana S Guerreiro Stücklin, Nicolas U Gerber, André O von Bueren, Claudia Khurana, Norbert Jorch, Maria Wiese, Christian P Kratz, Matthias Eyrich, Michael Karremann, Ulrich Herrlinger, Michael Hölzel, David T W Jones, Marion Hoffmann, Torsten Pietsch, Gerrit H Gielen, Christof M Kramm

1. FEBRUAR 2025 (GAIN)

Bruton's tyrosine kinase (BTK) and matrix metalloproteinase-9 (MMP-9) regulate NLRP3 inflammasome-dependent cytokine and neutrophil extracellular trap responses in primary neutrophils

J Allergy Clin Immunol. 2025 Feb. >>[Pubmed-Link](#)<<

Vinicius N C Leal, Francesca Bork, Maria Mateo Tortola, Juli-Christin von Guillaume, Carsten L Greve, Stefanie Bugl, Bettina Danker, Zsafia A Bittner, Bodo Grimbacher, Alessandra Pontillo, Alexander N R Weber

30. JANUAR 2025 (MYPRED)

Genetic and clinical spectrum of SAMD9 and SAMD9L syndromes: from variant interpretation to patient management

Blood. 2025 Jan 30. >>[Pubmed-Link](#)<<

Sushree S Sahoo, Miriam Erlacher, Marcin W Wlodarski

28. JANUAR 2025 (TREATHSP.NET)

NPT100-18A rescues mitochondrial oxidative stress and neuronal degeneration in human iPSC-based Parkinson's model

BMC Neurosci. 2025 Jan 28. >>[Pubmed-Link](#)<<

Julian E Alecu, Veronika Sigutova, Razvan-Marius Brazdis, Sandra Lörentz, Marios Evangelos Bogiongko, Anara Nursaitova, Martin Regensburger, Laurent Roybon, Kerstin M Galler, Wolfgang Wrasidlo, Beate Winner, Iryna Prots

28. JANUAR 2025 (HICHOL)

Comparison of different microbiome analysis pipelines to validate their reproducibility of gastric mucosal microbiome composition

mSystems. 2025 Jan 28. >>[Pubmed-Link](#)<<

Konrad Lehr, Baptiste Oosterlinck, Chee Kin Then, Matthew R Gemmell, Rolandas Gedgaudas, Jan Bornschein, Juozas Kupcinskas, Annemieke Smet, Georgina Hold, Alexander Link; ENIGMA: European Network for the Investigation of Gastrointestinal Mucosal Alterations

28. JANUAR 2025 (ADDRESS)

Cancer risk in carriers of TP53 germline variants grouped into different functional categories

JNCI Cancer Spectr. 2025 Jan 28. >>[Pubmed-Link](#)<<

Lucas John Müntnich, Christina M Dutzmann, Anika Großhennig, Valentina Härter, Myriam Keymling, Angela Mastronuzzi, Emilie Montellier, Juliane Nees, Natalie E Palmaers, Judith Penkert, Stefan M Pfister, Tim Ripperger, Sarah Schott, Farina Silchmüller, Pierre Hainaut, Christian P Kratz

23. JANUAR 2025 (STOP-FSGS)

Characterizing and targeting glioblastoma neuron-tumor networks with retrograde tracing

Cell. 2025 Jan 23. >>[Pubmed-Link](#)<<

Svenja K Tetzlaff, Ekin Reyhan, Nikolas Layer, C Peter Bengtson, Alina Heuer, Julian Schroers, Anton J Faymonville, Atefeh Pourkhalili Langeroudi, Nina Drewa, Elijah Keifert, Julia Wagner, Stella J Soyka, Marc C Schubert, Niroshan Sivapalan, Rangel L Pramatarov, Verena Buchert, Tim Wageringel, Elena Grabis, Niklas Wißmann, Obada T Alhalabi, Michael Botz, Jovana Bojceviski, Joaquín Campos, Berin Boztepe, Jonas G Scheck, Sascha Henry Conic, Maria C Puschhof, Giulia Villa, Richard Drexler, Yahya Zghaibeh, Fabian Hausmann, Sonja Hänzelmann, Matthias A Karreman, Felix T Kurz, Manuel Schröter, Marc Thier, Abigail K Suwala, Karin Forsberg-Nilsson, Claudio Acuna, Julio Saez-Rodriguez, Amir Abdollahi, Felix Sahn, Michael O Breckwoldt, Bogdana Suchorska, Franz L Ricklefs, Dieter Henrik Heiland, Varun Venkataramani

8. JANUAR 2025 (TREATHSP.NET)

White matter aging and its impact on brain function

Neuron. 2025 Jan 8. >>[Pubmed-Link](#)<<

Janos Groh, Mikael Simons

6. JANUAR 2025 (GAIN)

The effect of HLA genotype on disease onset and severity in CTLA-4 insufficiency

Front Immunol. 2025 Jan 6. eCollection 2024. >>[Pubmed-Link](#)<<

Sara Posadas-Cantera, Noriko Mitsuiki, Florian Emmerich, Virginia Patiño, Hanns-Martin Lorenz, Olaf Neth, Ingunn Dybedal, Kjetil Taskén, Alejandro A Schäffer, Bodo Grimbacher, Laura Gámez-Díaz

1. JANUAR 2025 (NEOCYST)

Urinary Dickkopf-3 Reflects Disease Severity and Predicts Short-Term Kidney Function Decline in Renal Ciliopathies

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

Mareike Dahmer-Heath, Joachim Gerß, Danilo Fliser, Max Christoph Liebau, Thimoteus Speer, Anna-Katharina Telgmann, Kathrin Burgmaier, Petra Pennekamp, Lars Pape, Franz Schaefer, Martin Konrad, Jens Christian König, NEOCYST Consortium

1. JANUAR 2025 (CONNECT-GENERATE)

Spectrum of Clinical and Imaging Features of Children With GFAP Astrocytopathy

Neurol Neuroimmunol Neuroinflamm. 2025 Jan. >>[Pubmed-Link](#)<<

Simon Sommer, Andreas Panzer, Annikki Bertolini, Robert Cleaveland, Vivek Jain, Tugba Kapanci, Ute Derichs, Tobias Geis, Axel Neu, Christa Löhr-Nilles, Rahel Aeschmann-Huhn, Marina Flotats-Bastardas, Kumaran Deiva, Thais Armangue, Gemma Olivé-Girera, Sudheeran Kanno, Anne Koy, Hadas Meirson, Aviva Fattal-Valevski, Esther Ganelin-Cohen, Heike Losch, Annacarin Horne, Ronny Wickström, Justina Dargvainiene, Frank Leypoldt, Kevin Rostasy

1. JANUAR 2025 (CONNECT-GENERATE)

Patient-Reported Outcome Measures in NMDA Receptor Encephalitis

Neurol Neuroimmunol Neuroinflamm. 2025 Jan. Epub 2024 Dec 12. >>[Pubmed-Link](#)<<

Josephine Heine, Ole Jonas Boeken, Sophia Rekers, Katharina Wurdack, Harald Prüss, Carsten Finke

1. JANUAR 2025 (CONNECT-GENERATE)

Amphiphysin and GAD65 IgG antibodies in patients with obsessive-compulsive syndromes

Eur Neuropsychopharmacol. 2025 Jan. Epub 2024 Nov 30. >>[Pubmed-Link](#)<< (Letter)

Katharina von Zedtwitz, Bernd Feige, Alexander Maier, Sarah L Schaefer, Kathrin Nickel, Marco Reisert, Kai Spiegelhalder, Nils Venhoff, Joachim Brumberg, Horst Urbach, Rick Dersch, Miriam A Schiele, Katharina Domschke, Ludger Tebartz van Elst, Volker A Coenen, Luciana Hannibal, Harald Prüss, Simon J Maier, Dominique Endres

1. JANUAR 2025 (GAIN)

Type-Specific Impacts of Protein Defects in Pathogenic NFKB2 Variants: Novel Clinical Findings From 138 Patients

J Allergy Clin Immunol Pract. 2025 Jan. >>[Pubmed-Link](#)<<

Jan Meissner, Manfred Fliegau, Bodo Grimbacher, Christian Klemann