

## Veröffentlichungen der BMBF-geförderten Forschungsverbände für seltene Erkrankungen, 2023

### 20. DEZEMBER 2023 (TREATHSP.NET)

#### **Novel Homozygous FA2H Variant Causing the Full Spectrum of Fatty Acid Hydroxylase-Associated Neurodegeneration (SPG35)**

Genes (Basel). 2023 Dec 20. >>[PubMed-Link](#)<<

Alexander German, Jelena Jukic, Andreas Laner, Philipp Arnold, Eileen Socher, Angelika Mennecke, Manuel A Schmidt, Jürgen Winkler, Angela Abicht, Martin Regensburger

### 19. DEZEMBER 2023 (STOP-FSGS)

#### **In vivo characterization of a podocyte-expressed short podocin isoform**

BMC Nephrol. 2023 Dec 19. >>[PubMed-Link](#)<<

Linus Butt, David Unnersjö-Jess, Dervla Reilly, Robert Hahnfeldt, Markus M Rinschen, Katarzyna Bozek, Bernhard Schermer, Thomas Benzing, Martin Höhne

### 13. DEZEMBER 2023 (STOP-FSGS)

#### **Research on Rare Diseases in Germany – Using small fish and super-resolution microscopy to track down a rare disease**

J Health Monit. 2023 Dec 13. >>[PubMed-Link](#)<<

Deutsche Fassung: **Forschung zu Seltenen Erkrankungen in Deutschland – Mit kleinen Fischen und Superresolution-Mikroskopie auf den Spuren einer seltenen Erkrankung**

Nicole Endlich

### 13. DEZEMBER 2023 (GAIN)

#### **Research on Rare Diseases in Germany – The GAIN Registry: a registry for individuals with congenital multi-organ autoimmune diseases**

J Health Monit. 2023 Dec 13. >>[PubMed-Link](#)<<

Deutsche Fassung: **Forschung zu Seltenen Erkrankungen in Deutschland – Das GAIN-Register: Ein Register für Personen mit angeborenen Multi-Organ-Autoimmunerkrankungen**

Cynthia Stapornwongkul, Alexandra Nieters, Paulina Staus, Stephan Rusch, Anita Delor, Ulrich Baumann, Julius Wehrle, Melanie Boerries, Markus G Seidel, Bodo Grimbacher, Gerhard Kindle

### 13. DEZEMBER 2023 (ADDRESS)

#### **Research on Rare Diseases in Germany – The cancer predisposition syndrome registry**

J Health Monit. 2023 Dec 13. >>[PubMed-Link](#)<<

Deutsche Fassung: **Forschung zu Seltenen Erkrankungen in Deutschland – Das Krebsprädispositionssyndrom-Register**

Christina M Dutzmann, Nathalie E Palmaers, Lucas J Müntnich, Farina J Strüwe, Judith Penkert, Birte Sängler, Beatrice Hoffmann, Anja Karow, Christina Reimer, Tanja Gerasimov, Marena R Niewisch, Christian P Kratz

### 7. DEZEMBER 2023 (GAIN)

#### **Rituximab to treat prolidase deficiency due to a novel pathogenic copy number variation in PEPD**

RMD Open. 2023 Dec 7. >>[PubMed-Link](#)<<

Faranaz Atschekzei, Mykola Fedchenko, Abdulwahab Elsayed, Natalia Dubrowinskaja, Theresa Graalmann, Felix C Ringshausen, Torsten Witte, Georgios Sogkas

### 1. DEZEMBER 2023 (TREAT-ION)

#### **SLC6A1 variant pathogenicity, molecular function and phenotype: a genetic and clinical analysis**

Brain. 2023 Dec 1. >>[PubMed-Link](#)<<

Arthur Stefanski, Eduardo Pérez-Palma, Tobias Brünger, Ludovica Montanucci, Cornelius Gati, Chiara Klöckner, Katrine M Johannesen, Kimberly Goodspeed, Marie Macnee, Alexander T Deng, Ángel Aledo-Serrano, Artem Borovikov, Maina Kava, Arjan M Bouman, M J Hajianpour, Deb K Pal, Marc Engelen, Eveline E O Hagebeuk, Marwan Shinawi, Alexis R Heidlebaugh, Kathryn Oetjens, Trevor L Hoffman, Pasquale Striano, Amanda S Freed, Line Futtrup, Thomas Balslev, Anna Abulí, Leslie Danvoye, Damien Lederer, Tugce Balci, Maryam Nabavi Nouri, Elizabeth Butler, Sarah Drewes, Kalene van Engelen, Katherine B Howell, Jean Houry, Patrick May, Marena Trinidad, Steven Froelich, Johannes R Lemke 5, Jacob Tiller, Amber N Freed, Jing-Qiong Kang, Arthur Wuster, Rikke S Møller, Dennis Lal

### 1. DEZEMBER 2023 (TREAT-ION)

#### **Clinical and electrophysiological features of SCN8A variants causing episodic or chronic ataxia**

eBioMedicine 2023;98:104855. >>[Journal-Link](#)<<

Hang Lyu, Christian M. Boßelmann, Katrine M. Johannesen, Mahmoud Koko, Juan Dario Ortigoza-Escobar, Sergio Aguilera-Albesa, Deyanira Garcia-Navas Núñez, Tarja Linnankivi, Eija Gaily, Henriette J. A. van Ruiten, Ruth Richardson, Cornelia Betzler, Gabriella Horvath, Eva Brilstra, Niels Geerdink, Daniele Orsucci, Alessandra Tessa, Elena Gardella, Zofia Fleszar, Ludger Schöls, Holger Lerche, Rikke S. Møller, Yuanyuan Liu

### 1. DEZEMBER 2023 (STOP-FSGS)

#### **A Novel High-Content Screening Assay Identified Belinostat as Protective in a FSGS-Like Zebrafish Model**

J Am Soc Nephrol. 2023 Dec 1. >>[PubMed-Link](#)<<

Maximilian Schindler, Florian Siegerist, Tim Lange, Stefan Simm, Sophia-Marie Bach, Marianne Klawitter, Jochen Gehrig, Sheraz Gul, Nicole Endlich

### 29. NOVEMBER 2023 (GAIN)

#### **GenIA, the Genetic Immunology Advisor database for Inborn Errors of Immunity**

J Allergy Clin Immunol. 2023 Nov 29 >>[PubMed-Link](#)<<

Andrés Caballero-Oteyza, Laura Crisponi, Xiao P Peng, Kevin Yauy, Stefano Volpi, Stefano Giardino, Alexandra F Freeman, Bodo Grimbacher, Michele Proietti

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### 28. NOVEMBER 2023 (STOP-FSGS)

#### **Evaluation of the efficacy of cystinosin supplementation through CTNS mRNA delivery in experimental models for cystinosis**

Sci Rep. 2023 Nov 283. >>[PubMed-Link](#)<<

Tjessa Bondue, Sante Princiero Berlingiero, Florian Siegerist, Elena Sendino-Garví, Maximilian Schindler, Hans Jacobus Baelde, Sara Cairoli, Bianca Maria Goffredo, Fanny Oliveira Arcolino, Jürgen Dieker, Manoe Jacoba Janssen, Nicole Endlich, Roland Brock, Rik Gijsbers, Lambertus van den Heuvel, Elena Levtchenko

### 26. NOVEMBER 2023 (MYPRED)

#### **Development of MDS in Pediatric Patients with GATA2 Deficiency: Increased Histone Trimethylation and Deregulated Apoptosis as Potential Drivers of Transformation**

Cancers (Basel). 2023 Nov 26. >>[PubMed-Link](#)<<

Franziska Schreiber, Guido Piontek, Yuki Schneider-Kimoto, Stephan Schwarz-Furlan, Rita De Vito, Franco Locatelli, Carole Gengler, Ayami Yoshimi, Andreas Jung, Frederick Klauschen, Charlotte M Niemeyer, Miriam Erlacher, Martina Rudelius

### 23. NOVEMBER 2023 (STOP-FSGS)

#### **3D Printed Tubulointerstitium Chip as an In Vitro Testing Platform**

Macromol Biosci. 2023 Nov 23. >>[PubMed-Link](#)<<

Gabriele Addario, Daphne Eussen, Sonja Djudjaj, Peter Boor, Lorenzo Moroni, Carlos Mota

### 14. NOVEMBER 2023 (MITONET)

#### **Dystonia in ATP Synthase Defects: Reconnecting Mitochondria and Dopamine**

Mov Disord. 2023 Nov 14. >>[PubMed-Link](#)<<

Elisabetta Indelicato, Sylvia Boesch, Niccolò E Mencacci, Daniele Ghezzi, Holger Prokisch, Juliane Winkelmann, Michael Zech

### 8. NOVEMBER 2023 (GAIN)

#### **Autoantibodies against type I IFNs in humans with alternative NF-κB pathway deficiency**

Nature. 2023 Nov. >>[PubMed-Link](#)<<

Tom Le Voyer, Audrey V Parent, Xian Liu et al.

### 8. NOVEMBER 2023 (GAIN)

#### **Telomere biology disorders may manifest as common variable immunodeficiency (CVID)**

Clin Immunol. 2023 Nov 8. >>[PubMed-Link](#)<<

Benjamin Rolles, Andres Caballero-Oteyza, Michele Proietti, Sigune Goldacker, Klaus Warnatz, Nadezhda Camacho-Ordóñez, Seraina Prader, Jana Pachlopnik Schmid, Margherita Vieri, Susanne Isfort, Robert Meyer, Martin Kirschner, Tim H Brümmendorf, Fabian Beier, Bodo Grimbacher

#### 7. NOVEMBER 2023 (ADDRESS)

##### **Medical students' knowledge on cancer predisposition syndromes and attitude toward eHealth**

Arch Gynecol Obstet. 2023 Nov 7. >>[PubMed-Link](#)<<

Juliane Nees, Farina Struewe, Sarah Schott

#### 6. NOVEMBER 2023 (NEOCYST)

##### **Kif21a deficiency leads to impaired glomerular filtration barrier function**

Sci Rep. 2023 Nov 6. >>[PubMed-Link](#)<<

Hanna Riedmann, Séverine Kayser, Martin Helmstädter, Daniel Epting, Carsten Bergmann

#### 5. NOVEMBER 2023 (GAIN)

##### **JAK inhibitor treatment for inborn errors of JAK/STAT signaling: An ESID/EBMT-IEWP retrospective study**

J Allergy Clin Immunol. 2024 Jan. Epub 2023 Nov 5. >>[PubMed-Link](#)<<

Marco Fischer, Peter Olbrich, Jérôme Hadjadj, Volker Aumann, Shahrzad Bakhtiar, Vincent Barlogis, Philipp von Bismarck, Markéta Bloomfield, Claire Booth, Emmeline P Buddingh, Deniz Cagdas, Martin Castelle, Alice Y Chan, Shanmuganathan Chandrakasan, Kritika Chetty, Pierre Cougoul, Etienne Crickx, Jasmeen Dara, Angela Deyà-Martínez, Susan Farmand, Renata Formankova, Andrew R Genney, Luis Ignacio Gonzalez-Granado, David Hagin, Leif Gunnar Hanitsch, Jana Hanzliková, Fabian Hauck, José Ivorra-Cortés, Kai Kisand, Ayca Kiykim, Julia Köhrholz, Timothy Ronan Leahy, Joris van Montfrans, Zohreh Nademi, Brigitte Nelken, Suhag Parikh, Silvi Plado, Jan Ramakers, Antje Redlich, Frédéric Rieux-Laucat, Jacques G Rivière, Yulia Rodina, Pérsio Roxo Júnior, Sarah Salou, Catharina Schuetz, Anna Shcherbina, Mary A Slatter, Fabien Touzot, Ekrem Unal, Arjan C Lankester, Siobhan Burns, Mikko R J Seppänen, Olaf Neth, Michael H Albert, Stephan Ehl, Bénédicte Neven, Carsten Speckmann

#### 1. NOVEMBER 2023 (STOP-FSGS)

##### **Introduction of a novel chimeric active immunization mouse model of PLA2R1-associated membranous nephropathy**

Kidney Int. 2023 Nov. >>[PubMed-Link](#)<<

Nicola M Tomas, Annabel Schnarre, Silke Dehde, Renke Lucas, Irm Hermans-Borgmeyer, Oliver Kretz, Sarah M S Koellner, Thorsten Wiech, Friedrich Koch-Nolte, Larissa Seifert, Tobias B Huber, Gunther Zahner

#### 1. NOVEMBER 2023 (STOP-FSGS)

##### **JNKs protect from cholestatic liver disease progression by modulating Apelin signalling**

JHEP Rep. 2023 Jul 18. eCollection 2023 Nov. >>[PubMed-Link](#)<<

Mohamed Ramadan Mohamed, Johannes Haybaeck, Hanghang Wu, Huan Su, Matthias Bartneck, Cheng Lin, Mark V Boeschoten, Peter Boor, Benjamin Goeppert, Christian Rupp, Pavel Strnad, Roger J Davis, Francisco Javier Cubero, Christian Trautwein

#### 1. NOVEMBER 2023 (GAIN)

##### **Next generation sequencing (NGS)-based approach to diagnosing Algerian patients with suspected inborn errors of immunity (IEIs)**

Clin Immunol. 2023 Nov. >>[PubMed-Link](#)<<

Xiao P Peng, Moudjahed Saleh Al-Ddafari, Andres Caballero-Oteyza, Chahrazed El Mezouar, Pavla Mrovecova, Saad Eddin Dib, Zoheir Massen, Mohammed Chems-Eddine Smahi, Alddafari Faiza, Rafik Terki Hassaïne, Gérard Lefranc, Mourad Aribi, Bodo Grimbacher

#### 1. NOVEMBER 2023 (GENERARE)

##### **NECAB2 is an endosomal protein important for striatal function**

Free Radic Biol Med. 2023 Nov 1. >>[PubMed-Link](#)<<

Diones Bueno, Partha Narayan Dey, Teresa Schacht, Christina Wolf, Verena Wüllner, Elena Morpurgo, Liliana Rojas-Charry, Lena Sessinghaus, Petra Leukel, Clemens Sommer, Konstantin Radyushkin, Luise Florin, Jan Baumgart, Paul Stamm, Andreas Daiber, Guilherme Horta, Leonardo Nardi, Verica Vasic, Michael J Schmeisser, Andrea Hellwig, Angela Oskamp, Andreas Bauer, Ruchika Anand, Andreas S Reichert, Sandra Ritz, Gianluigi Nocera, Claire Jacob, Jonas Peper, Marion Silies, Katrin B M Frauenknecht, Michael K E Schäfer, Axel Methner

#### 31. OKTOBER 2023 (TREATHSP.NET)

##### **The kinesin motor KIF1C is a putative transporter of the exon junction complex in neuronal cells**

RNA. 2022 Oct 31. >>[PubMed-Link](#)<<

Maike Nagel, Marvin Noss, Jishu Xu, Nicola Horn, Marius Ueffing, Karsten Boldt, Rebecca Schüle

**31. OKTOBER 2023 (CONNECT-GENERATE)****Human NMDAR autoantibodies disrupt excitatory-inhibitory balance, leading to hippocampal network hypersynchrony**Cell Rep. 2023 Oct 31. >>[PubMed-Link](#)<<

Mihai Ceanga, Vahid Rahmati, Holger Haselmann, Lars Schmidl, Daniel Hunter, Anna-Katherina Brauer, Sabine Liebscher, Jakob Kreye, Harald Prüss, Laurent Groc, Stefan Hallermann, Josep Dalmau, Alessandro Ori, Manfred Heckmann, Christian Geis

**24. OKTOBER 2023 (MYPRED)****Somatic mutational landscape of hereditary hematopoietic malignancies caused by germline variants in RUNX1, GATA2, and DDX41**Blood Adv. 2023 Oct 24. >>[PubMed-Link](#)<<

Claire C Homan, Michael W Drazer, Kai Yu, David M Lawrence, Jinghua Feng, Luis Arriola-Martinez, Matthew J Pozsgai, Kelsey E McNeely, Thuong Ha, Parvathy Venugopal, Peer Arts, Sarah L King-Smith, Jesse Cheah, Mark Armstrong, Paul Wang, Csaba Bödör, Alan B Cantor, Mario Cazzola, Erin Degelman, Courtney D DiNardo, Nicolas Duployez, Remi Favier, Stefan Fröhling, Ana Rio-Machin, Jeffery M Klco, Alwin Krämer, Mineo Kurokawa, Joanne Lee, Luca Malcovati, Neil V Morgan, Georges Natsoulis, Carolyn Owen, Keyur P Patel, Claude Preudhomme, Hana Raslova, Hugh Rienhoff, Tim Ripperger, Rachael Schulte, Kiran Tawana, Elvira Velloso, Benedict Yan, Erika Kim, Raman Sood, Amy P Hsu, Steven M Holland, Kerry Phillips, Nicola K Poplawski, Milena Babic, Andrew H Wei, Cecily Forsyth, Helen Mar Fan, Ian D Lewis, Julian Cooney, Rachel Susman, Lucy C Fox, Piers Blombery, Deepak Singhal, Devendra Hiwase, Belinda Phipson, Andreas W Schreiber, Christopher N Hahn, Hamish S Scott, Paul Liu, Lucy A Godley, Anna L Brown; NISC Comparative Sequencing Program

**24. OKTOBER 2023 (CONNECT-GENERATE)****Impaired functional connectivity of the hippocampus in translational murine models of NMDA-receptor antibody associated neuropsychiatric pathology**Mol Psychiatry. 2023 Oct 24. >>[PubMed-Link](#)<<

Joseph Kuchling, Betty Jurek, Mariya Kents, Jakob Kreye, Christian Geis, Jonathan Wickel, Susanne Mueller, Stefan Paul Koch, Philipp Boehm-Sturm, Harald Prüss, Carsten Finker

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**17. OKTOBER 2023 (NEOCYST)****A novel role for the chloride intracellular channel protein Clic5 in ciliary function**Sci Rep. 2023 Oct 17. >>[PubMed-Link](#)<<

Elisabeth Ott, Sylvia Hoff, Lara Indorf, Franck Anicet Ditengou, Julius Müller, Gina Renschler, Soeren S Lienkamp, Albrecht Kramer-Zucker, Carsten Bergmann, Daniel Epting

**16. OKTOBER 2023 (STOP-FSGS)****Banff Digital Pathology Working Group: Image Bank, Artificial Intelligence Algorithm, and Challenge Trial Developments**Transpl Int. 2023 Oct 16. >>[PubMed-Link](#)<<

Alton B Farris, Mariam P Alexander, Ulysses G J Balis, Laura Barisoni, Peter Boor, Roman D Bülow, Lynn D Cornell, Anthony J Demetris, Evan Farkash, Meyke Hermsen, Julien Hogan, Renate Kain, Jesper Kers, Jun Kong, Richard M Levenson, Alexandre Loupy, Maarten Naesens, Pinaki Sarder, John E Tomaszewski, Jeroen van der Laak, Dominique van Midden, Yukako Yagi, Kim Solez

**12. OKTOBER 2023 (GENERARE)****Epilepsy in cardiofaciocutaneous syndrome: Clinical burden and response to anti-seizure medication**Am J Med Genet A. 2023 Oct 12. >>[PubMed-Link](#)<<

Daniel L Kenney-Jung, Josue E Collazo-Lopez, Dante J Rogers, Ryan Shanley, Abigail L Zatkalik, Ashley E Whitmarsh, Amy E Roberts, Martin Zenker, Elizabeth I Pierpont

**11. OKTOBER 2023 (TREAT-ION)****Voltage-gated calcium channels in genetic epilepsies**J Neurochem. 2023 Oct 11. >>[PubMed-Link](#)<<

Robert J Lauerer, Holger Lerche



### 9. OKTOBER 2023 (HICHOLO)

#### **Insulin determines TGF- $\beta$ effects on HNF4 $\alpha$ transcription in hepatocytes**

Am J Pathol. 2023 Oct 9. >>[PubMed-Link](#)<<

Rilu Feng, Chenhao Tong, Tao Lin 1, Hui Liu, Chen Shao, Yujia Li, Carsten Sticht, Kejia Kan, Xiaofeng Li, Rui Liu, Sai Wang, Shanshan Wang, Stefan Munker, Hanno Niess, Christoph Meyer, Roman Liebe, Matthias P Ebert, Steven Dooley, Hua Wang, Huiguo Ding, Hong-Lei Weng

### 3. OKTOBER 2023 (TREATHSP.NET)

#### **SARM1 deletion delays cerebellar but not spinal cord degeneration in an enhanced mouse model of SPG7 deficiency**

Brain. 2023 Oct 3. >>[PubMed-Link](#)<<

Carolina Montoro-Gámez, Hendrik Nolte, Thibaut Molinié, Giovanna Evangelista, Simon E Tröder, Esther Barth, Milica Popovic, Aleksandra Trifunovic, Branko Zevnik, Thomas Langer, Elena I Rugarli

### 3. OKTOBER 2023 (ADDRESS)

#### **Multiple TP53 p.R337H haplotypes and implications for tumor susceptibility**

HGG Adv. 2023 Oct 3;5(1):100244. >>[PubMed-Link](#)<<

Emilia M Pinto, Cintia Fridman, Bonald C Figueiredo, Hector Salvador, Manuel R Teixeira, Carla Pinto, Manuela Pinheiro, Christian P Kratz, Cinzia Lavarino, Edith A M F Legal, Anh Le, Gregory Kelly, Erika Koeppel, Elena M Stoffel, Kelsey Breen, Stefanie Hahner, Britta Heinze, Piti Techavichit, Amanda Krause, Tsutomu Ogata, Yasuko Fujisawa, Michael F Walsh, Huma Q Rana, Kara N Maxwell, Judy E Garber, Carlos Rodriguez-Galindo, Raul C Ribeiro, Gerard P Zambetti

### 3. OKTOBER 2023 (GAIN)

#### **Precision medicine in monogenic inflammatory bowel disease: proposed mIBD REPORT standards**

Nat Rev Gastroenterol Hepatol. 2023 Oct 3. >>[PubMed-Link](#)<<

Holm H Uhlig, Claire Booth, Judy Cho, Marla Dubinsky, Anne M Griffiths, Bodo Grimbacher, Sophie Hambleton, Ying Huang, Kelsey Jones, Jochen Kammermeier, Hirokazu Kanegane, Sibylle Koletzko, Daniel Kotlarz, Christoph Klein, Michael J Lenardo, Bernice Lo, Dermot P B McGovern, Ahmet Özen, Lissy de Ridder, Frank Ruemmele, Dror S Shouval, Scott B Snapper, Simon P Travis, Dan Turner, David C Wilson, Aleixo M Muise

### 3. OKTOBER 2023 (CONNECT-GENERATE)

#### **Post-herpes simplex virus encephalitis autoimmunity: more the rule than the exception**

Brain. 2023 Oct 3. >>[PubMed-Link](#)<<

(Scientific Commentary)

Frank Leypoldt, Klaus-Peter Wandinger

### 1. OKTOBER 2023 (GAIN)

#### **Activated phosphoinositide 3-kinase $\delta$ syndrome: Update from the ESID Registry and comparison with other autoimmune-lymphoproliferative inborn errors of immunity**

J Allergy Clin Immunol. 2023 Oct. >>[PubMed-Link](#)<<

Maria Elena Maccari, Martin Wolkewitz, Charlotte Schwab, Tiziana Lorenzini, Jennifer W Leiding, Nathalie Aladjdi, Hassan Abolhassani, Wadih Abou-Chahla, Alessandro Aiuti, Saba Azarnoush, Safa Baris, Vincent Barlogis, Federica Barzaghi, Ulrich Baumann, Marketa Bloomfield, Nadezda Bohynikova, Damien Bodet, David Boutboul, Giorgia Bucciol, Matthew S Buckland, Siobhan O Burns, Caterina Cancrini, Pascal Cathébras, Marina Cavazzana, Morgane Cheminant, Matteo Chinello, Peter Cizna, Tanya I Coulter, Maud D'Aveni, Olov Ekwall, Zelimir Eric, Efreem Eren, Anders Fasth, Pierre Frange, Benjamin Fournier, Marina Garcia-Prat, Martine Gardembas, Christoph Geier, Sujal Ghosh, Vera Goda, Lennart Hammarström, Fabian Hauck, Maximilian Heeg, Edyta Heropolitanska-Pliszka, Anna Hilfanova, Stephen Jolles, Elif Karakoc-Aydiner, Gerhard R Kindle, Ayca Kiykim, Christian Klemann, Patra Koletsi, Sylwia Koltan, Irina Kondratenko, Julia Körholz, Renate Krüger, Eric Jeziorski, Romain Levy, Guillaume Le Guenno, Guillaume Lefevre, Vassilios Lougaris, Antonio Marzollo, Nizar Mahlaoui, Marion Malphettes, Andrea Meinhardt, Etienne Merlin, Isabelle Meyts, Tomas Milota, Fernando Moreira, Despina Moshous, Anna Mukhina, Olaf Neth, Jennifer Neubert, Benedicte Neven, Alexandra Nieters, Raphael Nove-Josserand, Eric Oksenhendler, Ahmet Ozen, Peter Olbrich, Antoinette Perlat, Malgorzata Pac, Jana Pachlopnik Schmid, Lucia Pacillo, Alba Parra-Martinez, Olga Paschenko, Isabelle Pellier, Asena Pinar Sefer, Alessandro Plebani, Dominique Plantaz, Seraina Prader, Loic Raffray, Henrike Ritterbusch, Jacques G Riviere, Beatrice Rivalta, Stephan Rusch, Inga Sakovich, Sinisa Savic, Raphael Scheible, Nicolas Schleinitz, Catharina Schuetz, Ansgar Schulz, Anna Sediva, Michaela Semeraro, Svetlana O Sharapova, Anna Shcherbina, Mary A Slatter, Georgios Sogkas, Pere Soler-Palacin, Carsten Speckmann, Jean-Louis Stephan, Felipe Suarez, Alberto Tommasini, Johannes Trück, Annette Uhlmann, Koen J van Aerde, Joris van Montfrans, Horst von Bernuth,

Klaus Warnatz, Tony Williams, Austen J J Worth, Winnie Ip, Capucine Picard, Emilie Catherinot, Zohreh Nademi, Bodo Grimbacher, Lisa R Forbes Satter, Sven Kracker, Anita Chandra, Alison M Condliffe, Stephan Ehl; European Society for Immunodeficiencies Registry Working Party

1. OKTOBER 2023 (MYPRED)

**Down syndrome and leukemia: from basic mechanisms to clinical advances**

Haematologica. 2023 Oct 1. >>[PubMed-Link](#)<<

André Baruchel, Jean-Pierre Bourquin, John Crispino, Sergi Cuartero, Henrik Hasle, Johann Hitzler, Jan-Henning Klusmann, Shai Izraeli, Andrew A Lane, Sébastien Malinge, Karen R Rabin, Irene Roberts, Sandra Ryeom, Sarah K Tasian, Elvin Wagenblast

1. OKTOBER 2023 (STOP-FSGS)

**Kidney fibrosis: Emerging diagnostic and therapeutic strategies**

Mol Aspects Med. 2023 Oct. >>[PubMed-Link](#)<<

Barbara Mara Klinkhammer, Peter Boor

25. SEPTEMBER 2023 (NEOCYST)

**Emerging principles of primary cilia dynamics in controlling tissue organization and function**

EMBO J. 2023 Sep 25. >>[PubMed-Link](#)<<

Jay Gopalakrishnan, Kerstin Feistel, Benjamin M Friedrich, Anne Grapin-Botton, Nathalie Jurisch-Yaksi, Elvira Mass, David U Mick, Roman-Ulrich Müller, Helen May-Simera, Bernhard Schermer, Miriam Schmidts, Peter Walentek, Dagmar Wachten

23. SEPTEMBER 2023 (GAIN)

**Functional Relevance of CTLA4 Variants: an Upgraded Approach to Assess CTLA4-Dependent Transendocytosis by Flow Cytometry**

J Clin Immunol. 2023 Sep 23. >>[PubMed-Link](#)<<

Jessica Rojas-Restrepo, Elena Sindram, Simon Zenke, Hanna Haberstroh, Noriko Mitsuiki, Annemarie Gabrysch, Katrin Huebscher, Sara Posadas-Cantera, Máté Krausz, Robin Kobbe, Jan C Rohr, Bodo Grimbacher, Laura Gámez-Díaz

22. SEPTEMBER 2023 (MYPRED)

**C-terminal variants in CDC42 drive type I interferon-dependent autoinflammation in NOCARH syndrome reversible by ruxolitinib**

Clin Immunol. 2023 Sep 22. >>[PubMed-Link](#)<<

Friedrich G Kapp, Stefanie Kretschmer, Cora C A Beckmann, Lena Wäsch, Anne Molitor, Raphaël Carapito, Mario Schubert, Nadja Lucas, Solène Conrad, Sylvaine Poignant, Bertrand Isidor, Meino Rohlf, Ayşenur Paç Kisaarslan, Denny Schanze, Martin Zenker, Annette Schmitt-Graeff, Brigitte Strahm, Anke Peters, Ayami Yoshimi, Wolfgang Driever, Thomas Zillinger, Claudia Günther, Shovamayee Maharana, Kaomei Guan, Christoph Klein, Stephan Ehl, Charlotte M Niemeyer, Ekrem Unal, Seiamak Bahram, Fabian Hauck, Min Ae Lee-Kirsch, Carsten Speckmann

22. SEPTEMBER 2023 (GAIN)

**C-terminal variants in CDC42 drive type I interferon-dependent autoinflammation in NOCARH syndrome reversible by ruxolitinib**

Clin Immunol. 2023 Sep 22. >>[PubMed-Link](#)<<

Friedrich G Kapp, Stefanie Kretschmer, Cora C A Beckmann, Lena Wäsch, Anne Molitor, Raphaël Carapito, Mario Schubert, Nadja Lucas, Solène Conrad, Sylvaine Poignant, Bertrand Isidor, Meino Rohlf, Ayşenur Paç Kisaarslan, Denny Schanze, Martin Zenker, Annette Schmitt-Graeff, Brigitte Strahm, Anke Peters, Ayami Yoshimi, Wolfgang Driever, Thomas Zillinger, Claudia Günther, Shovamayee Maharana, Kaomei Guan, Christoph Klein, Stephan Ehl, Charlotte M Niemeyer, Ekrem Unal, Seiamak Bahram, Fabian Hauck, Min Ae Lee-Kirsch, Carsten Speckmann

21. SEPTEMBER 2023 (GAIN)

**Case Report: Response of cutaneous lupus lesions in SLE to interferon receptor blockade parallels reduction of interferon score in blood**

Front Immunol. 2023 Sep 21. >>[PubMed-Link](#)<<

Claudia Günther, Christine Wolf, Louisa Fennen, Sarah Rösing, Stefan Beissert, Martin Aringer, Min Ae Lee-Kirsch

#### 20. SEPTEMBER 2023 (STOP-FSGS)

##### **DISCERN: deep single-cell expression reconstruction for improved cell clustering and cell subtype and state detection**

Genome Biol. 2023 Sep 20. >>[PubMed-Link](#)<<

Fabian Hausmann, Can Ergen, Robin Khatri, Mohamed Marouf, Sonja Hänzelmann, Nicola Gagliani, Samuel Huber, Pierre Machart, Stefan Bonn

#### 7. SEPTEMBER 2023 (GENERARE)

##### **Atrial fibrillation-associated electrical remodelling in human induced pluripotent stem cell-derived atrial cardiomyocytes: a novel pathway for antiarrhythmic therapy development**

Cardiovasc Res. 2023 Sep 7. >>[PubMed-Link](#)<<

Fitzwilliam Seibert, Tony Rubio, Robin Springer, Fiona Popp, Melanie Ritter, Aiste Liutkute, Lena Bartelt, Lea Stelzer, Fereshteh Haghighi, Jan Pietras, Hendrik Windel, Núria Díaz I Pedrosa, Markus Rapedius, Yannic Döring, Richard Solano, Robin Hindmarsh, Runzhu Shi, Malte Tiburcy, Tobias Brüggemann, Ingo Kutschka, Katrin Streckfuss-Bömeke, George Kensah, Lukas Cyganek, Wolfram H Zimmermann, Niels Voigt

#### 1. SEPTEMBER 2023 (MYPRED)

##### **Epigenome profiling reveals aberrant DNA methylation signature in GATA2 deficiency**

Haematologica. 2023 Sep 1. >>[PubMed-Link](#)<<

Oskar Marin-Bejar, Damia Romero-Moya, Javier Rodriguez-Ubrea, Maximiliano Distefano, Francesca Lessi, Paolo Aretini, Alessandro Liquori, Julio Castaño, Emilia Kozyra, Lili Kotmayer, Clara Bueno, José Cervera, José Carlos Rodríguez-Gallego, Josep F Nomdedeu, Laura Murillo-Sanjuán, Cristina Díaz De Heredia, Antonio Pérez-Martínez, Félix López-Cadenas, Carolina Martínez-Laperche, Nieves Dorado-Herrero, Francisco M Marco, Felipe Prósper, Pablo Menendez, David Valcárcel, Esteban Ballestar, Csaba Bödör, Anna Bigas, Albert Catalá, Marcin W Wlodarski, Alessandra Giorgetti

#### 1. SEPTEMBER 2023 (NEOCYST)

##### **Autosomal Recessive Polycystic Kidney Disease: Diagnosis, Prognosis, and Management**

Adv Kidney Dis Health. 2023 Sep. >>[PubMed-Link](#)<<

Kathrin Burgmaier, Ilse J Broekaert, Max C Liebau

#### 29. AUGUST 2023 (CONNECT-GENERATE)

##### **Synapsin autoantibodies during pregnancy are associated with fetal abnormalities**

Am J Pathol. 2023 Oct 9. >>[PubMed-Link](#)<<

Isabel Bünger, Ivan Talucci 3, Jakob Kreye, Markus Höltje, Konstantin L Makridis, Helle Foverskov Rasmussen, Scott van Hoof, César Cordero-Gomez, Tim Ullrich, Eva Sedlin, Kai Oliver Kreissner, Christian Hoffmann, Dragomir Milovanovic, Paul Turko, Friedemann Paul, Jessica Meckies, Stefan Verlohren, Wolfgang Henrich, Rabih Chaoui, Hans Michael Maric, Angela M Kaindl, Harald Prüss

#### 29. AUGUST 2023 (TREATHSP.NET)

##### **Automated assessment of foot elevation in adults with hereditary spastic paraplegia using inertial measurements and machine learning**

Orphanet J Rare Dis. 2023 Aug 29. >>[PubMed-Link](#)<<

Malte Ollenschläger, Patrick Höfner, Martin Ullrich, Felix Kluge, Teresa Greinwalder, Evelyn Loris, Martin Regensburger, Bjoern M Eskofier, Jürgen Winkler, Heiko Gaßner

#### 14. AUGUST 2023 (STOP-FSGS)

##### **An integrated organoid omics map extends modeling potential of kidney disease**

Nat Commun. 2023 Aug 14. >>[PubMed-Link](#)<<

Moritz Lassé, Jamal El Saghier, Celine C Berthier, Sean Eddy, Matthew Fischer, Sandra D Laufer, Dominik Kyllies, Arvid Hutzfeldt, Léna Lydie Bonin, Bernhard Dumoulin, Rajasree Menon, Virginia Vega-Warner, Felix Eichinger, Fadhl Alakwaa, Damian Fermin, Anja M Billing, Akihiro Minakawa, Phillip J McCown, Michael P Rose, Bradley Godfrey, Elisabeth Meister, Thorsten Wiech, Mercedes Noriega, Maria Chrysopoulou, Paul Brandts, Wenjun Ju, Linda Reinhard, Elion Hoxha, Florian Grahmmer, Maja T Lindenmeyer, Tobias B Huber, Hartmut Schlüter, Steffen Thiel, Laura H Mariani, Victor G Puelles, Fabian Braun, Matthias Kretzler, Fatih Demir, Jennifer L Harder, Markus M Rinschen

#### 7. AUGUST 2023 (STOP-FSGS)

##### **Extending the landscape of omics technologies by pathomics**

NPJ Syst Biol Appl. 2023. >>[PubMed-Link](#)<<

Roman D. Bülow, David L. Hölscher, Ivan G. Costa, Peter Boor

#### 5. AUGUST 2023 (GAIN)

##### **Hemophagocytic lymphohistiocytosis-like hyperinflammation due to a de novo mutation in DPP9**

J Allergy Clin Immunol. 2023 Aug 5. >>[PubMed-Link](#)<<

Christine Wolf, Hannah Fischer, Jörn-Sven Kühl, Sarah Koss, Rami Abou Jamra, Sven Starke, Jurek Schultz, Stephan Ehl, Katrin Neumann, Catharina Schuetz, Robert Huber, Veit Hornung, Min Ae Lee-Kirsch

#### 1. AUGUST 2023 (GAIN)

##### **Autoimmunity and immunodeficiency associated with monoallelic LIG4 mutations via haploinsufficiency**

J Allergy Clin Immunol. 2023 Aug. >>[PubMed-Link](#)<<

Annaïse J Jauch, Olivier Bignucolo, Sayuri Seki, Marie Ghraichy, Ottavia M Delmonte, Valentin von Niederhäusern, Rebecca Higgins, Adhideb Ghosh, Masako Nishizawa, Mariko Tanaka, Adrian Baldrich, Julius Köppen, Julia R Hirsiger, Robin Hupfer, Stephan Ehl, Anne Rensing-Ehl, Helmut Hopfer, Spasenija Savic Prince, Stephen R Daley, Florian A Marquardsen, Benedikt J Meyer, Michael Tamm, Thomas D Daikeler, Tamara Diesch, Thomas Kühne, Arthur Helbling, Caroline Berkemeier, Ingmar Heijnen, Alexander A Navarini, Johannes Trück, Jean-Pierre de Villartay, Annette Oxenius, Christoph T Berger, Christoph Hess, Luigi D Notarangelo, Hiroyuki Yamamoto, Mike Recher

#### 1. AUGUST 2023 (STOP-FSGS)

##### **2,8-Dihydroxyadenine-induced nephropathy causes hexosylceramide accumulation with increased mTOR signaling, reduced levels of protective SirT3 expression and impaired renal mitochondrial function**

Biochim Biophys Acta Mol Basis Dis. 2023 Aug 1. >>[PubMed-Link](#)<<

Julia Moellmann, Katja Krueger, Dickson W L Wong, Barbara M Klinkhammer, Eva M Buhl, Jonas Dehairs, Johan V Swinnen, Heidi Noels, Joachim Jankowski, Corinna Lebherz, Peter Boor, Nikolaus Marx, Michael Lehrke

#### 1. AUGUST 2023 (ADDRESS)

##### **MRD as Biomarker for Response to Donor Lymphocyte Infusion after Allogeneic Hematopoietic Cell Transplantation in Patients with AML**

Cancers (Basel). 2023 Aug 1 >>[PubMed-Link](#)<<

Katrin Teich, Michael Stadler, Razif Gabdoulline, Jyoti Kandarp, Clara Wienecke, Bennet Heida, Piroska Klement, Konstantin Büttner, Letizia Venturini, Martin Wichmann, Wolfram Puppe, Christian Schultze-Floreay, Christian Koenecke, Gernot Beutel, Matthias Eder, Arnold Ganser, Michael Heuser, Felicitas Thol

#### 1. AUGUST 2023 (GAIN)

##### **Clinical, immunological and molecular findings of 8 patients with typical and atypical severe combined immunodeficiency: identification of 7 novel mutations by whole exome sequencing**

Genes Immun. 2023 Aug. >>[PubMed-Link](#)<<

Zahra Alizadeh, Mohammad Reza Fazlollahi, Marzieh Mazinani, Mohsen Badalzadeh, Hanieh Heydarlou, Raphael Carapito, Anne Molitor, Andrés Caballero Garcia de Oteyza, Michele Proietti, Maryam Soleimani Bavani, Mansoureh Shariat, Morteza Fallahpour, Masoud Movahedi, Leila Moradi, Bodo Grimbacher, Seiamak Bahram, Zahra Pourpak

#### 23. JULI 2023 (MITONET)

##### **Dystonia Linked to EIF4A2 Haploinsufficiency: A Disorder of Protein Translation Dysfunction**

Mov Disord. 2023 Jul 23. >>[PubMed-Link](#)<<

Philip Harrer, Matej Škorvánek, Volker Kittke, Ivana Dzinovic, Friederike Borngreber, Mirja Thomsen, Vanessa Mandel, Tatiana Svorenova, Miriam Ostrozovicova, Kristina Kulcsarova, Riccardo Berutti, Hauke Busch, Fabian Ott, Robert Kopajtich, Holger Prokisch, Kishore R Kumar, Niccolo E Mencacci, Manju A Kurian, Alessio Di Fonzo, Sylvia Boesch, Andrea A Kühn, Ulrike Blümlein, Katja Lohmann, Bernhard Haslinger, David Weise, Robert Jech, Juliane Winkelmann, Michael Zech

#### 19. JULI 2023 (TREAT-ION)

##### **Apo state pore opening as functional basis of increased EAAT anion channel activity in episodic ataxia 6**

Front Physiol. 2023 Jul 19. >>[PubMed-Link](#)<<

Mariia Suslova, Daniel Kortzak, Jan-Philipp Machtens, Peter Kovermann, Christoph Fahlke



### 13. JULI 2023 (CONNECT-GENERATE)

#### **No evidence of neuronal/glial autoantibodies in febrile infection-related epilepsy syndrome (FIRES): a prospective clinic-serologic analysis**

Front Neurosci. 2023 Jul 13. >>[PubMed-Link](#)<<

Janina Soler Wenglein, Gerhard Kluger, Frank Leyboldt, Klaus-Peter Wandinger, Andreas van Baalen

### 6. JULI 2023 (STOP-FSGS)

#### **A YAP/TAZ-ARHGAP29-RhoA Signaling Axis Regulates Podocyte Protrusions and Integrin Adhesions**

Cells. 2023 Jul 6. >>[PubMed-Link](#)<<

Manuel Rogg, Jasmin I Maier, Martin Helmstädter, Alena Sammarco, Felix Kliewe, Oliver Kretz, Lisa Weißer, Clara Van Wymersch, Karla Findeisen, Anna L Koessinger, Olga Tsoy, Jan Baumbach, Markus Grabbert, Martin W

### 5. JULI 2023 (CONNECT-GENERATE)

#### **High frequency of cerebrospinal fluid autoantibodies in patients with seizures or epilepsies of unknown etiology**

Front Neurol. 2023 Jul 5. >>[PubMed-Link](#)<<

Paulina Schulz, Alva Lütt, Winfried Stöcker, Bianca Teegen, Martin Holtkamp, Harald Prüss

### 4. JULI 2023 (GENERARE)

#### **The Microenvironment of the Pathogenesis of Cardiac Hypertrophy**

Cells. 2023 Jul 4. >>[PubMed-Link](#)<<

Farhad Bazgir, Julia Nau, Saeideh Nakhaei-Rad, Ehsan Amin, Matthew J. Wolf, Jeffrey J. Saucerman, Kristina Lorenz, Mohammad Reza Ahmadian

### 4. JULI 2023 (GENERARE)

#### **ACE2-EGFR-MAPK signaling contributes to SARS-CoV-2 infection**

Life Sci. Alliance. 2023 Jul 4. >>[PubMed-Link](#)<<

Melanie Engler, Dan Albers, Pascal Von Maltitz, Rüdiger Groß, Jan Münch, Ion Cristian Cirstea

### 1. JULI 2023 (STOP-FSGS)

#### **Biallelic variants in CRIPT cause a Rothmund-Thomson-like syndrome with increased cellular senescence**

Genet Med. 2023 Jul. >>[PubMed-Link](#)<<

Luisa Averdunk, Maxim A Huetzen, Daniel Moreno-Andrés, Reinhard Kalb, Shane McKee, Tzung-Chien Hsieh, Annette Seibt, Marten Schouwink, Seema Lalani, Eissa Ali Faqeih, Theresa Brunet, Peter Boor, Kornelia Neveling, Alexander Hoischen, Barbara Hildebrandt, Elisabeth Graf, Linchao Lu, Weidong Jin, Joerg Schaper, Jamal A Omer, Tanguy Demaret, Nicole Fleischer, Detlev Schindler, Peter Krawitz, Ertan Mayatepek, Dagmar Wiczorek, Lisa L Wang, Wolfram Antonin, Ron D Jachimowicz, Verena von Felbert, Felix Distelmaier

### 1. JULI 2023 (GAIN)

#### **Sequencing the B Cell Receptor Repertoires of Antibody-Deficient Individuals With and Without Infection Susceptibility**

J Clin Immunol. 2023 Jul. >>[PubMed-Link](#)<<

Yoong Wearn Lim, Neftali Jose Ramirez, Michael A Asensio, Yao Chiang, Gabriele Müller, Pavla Mrovecova, Noriko Mitsuiki, Máté Krausz, Nadezhda Camacho-Ordóñez, Klaus Warnatz, Adam S Adler, Bodo Grimbacher

### 1. JULI 2023 (ADDRESS)

#### **Biallelic variants in CRIPT cause a Rothmund-Thomson-like syndrome with increased cellular senescence**

Genet Med. 2023 Jul. >>[PubMed-Link](#)<<

Luisa Averdunk, Maxim A Huetzen, Daniel Moreno-Andrés, Reinhard Kalb, Shane McKee, Tzung-Chien Hsieh, Annette Seibt, Marten Schouwink, Seema Lalani, Eissa Ali Faqeih, Theresa Brunet, Peter Boor, Kornelia Neveling, Alexander Hoischen, Barbara Hildebrandt, Elisabeth Graf, Linchao Lu, Weidong Jin, Joerg Schaper, Jamal A Omer, Tanguy Demaret, Nicole Fleischer, Detlev Schindler, Peter Krawitz, Ertan Mayatepek, Dagmar Wiczorek, Lisa L Wang, Wolfram Antonin, Ron D Jachimowicz, Verena von Felbert, Felix Distelmaier

### 27. JUNI 2023 (GAIN)

#### **Age-associated B cells predict impaired humoral immunity after COVID-19 vaccination in patients receiving immune checkpoint blockade**

Nat Commun. 2023 Jun 27. >>[PubMed-Link](#)<<

Juan Carlos Yam-Puc, Zhaleh Hosseini, Emily C Horner, Pehuén Pereyra Gerber, Nonantzin Beristain-Covarrubias, Robert Hughes, Aleksei Lulla, Maria Rust, Rebecca Boston, Magda Ali, Katrin Fischer, Edward

Simmons-Rosello, Martin O'Reilly, Harry Robson, Lucy H Booth, Lakmini Kahanawita, Andrea Correa-Noguera, David Favara, Lourdes Ceron-Gutierrez, Baerbel Keller, Andrew Craxton, Georgina S F Anderson, Xiao-Ming Sun, Anne Elmer, Caroline Saunders, Areti Bermperti, Sherly Jose, Nathalie Kingston, Thomas E Mulroney, Lucia P G Piñon; CITIID-NIHR COVID-19 BioResource Collaboration; Michael A Chapman, Sofia Grigoriadou, Marion MacFarlane, Anne E Willis, Kiran R Patil, Sarah Spencer, Emily Staples, Klaus Warnatz, Matthew S Buckland, Florian Hollfelder, Marko Hyvönen, Rainer Döffinger, Christine Parkinson, Sara Lear, Nicholas J Matheson, James E D Thaventhiran

#### 21. JUNI 2023 (GENERARE)

##### **Molecular and cellular evidence for the impact of a hypertrophic cardiomyopathy-associated RAF1 variant on the structure and function of contractile machinery in bioartificial cardiac tissues**

Commun Biol. 2023 Jun 21. >>[PubMed-Link](#)<<

Saeideh Nakhaei-Rad, Fereshteh Haghighi, Farhad Bazgir, Julia Dahlmann, Alexandra Viktoria Busley, Marcel Buchholzer, Karolin Kleemann, Anne Schänzer, Andrea Borchardt, Andreas Hahn, Sebastian Kötter, Denny Schanze, Ruchika Anand, Florian Funk, Annette Vera Kronenbitter, Jürgen Scheller, Roland P Piekorz, Andreas S Reichert, Marianne Volleth, Matthew J Wolf, Ion Cristian Cirstea, Bruce D Gelb, Marco Tartaglia, Joachim P Schmitt, Martina Krüger, Ingo Kutschka, Lukas Cyganek, Martin Zenker, George Kensah, Mohammad R Ahmadian

#### 20. JUNI 2023 (STOP-FSGS)

##### **Partner, Neighbor, Housekeeper and Dimension: 3D versus 2D Glomerular Co-Cultures Reveal Drawbacks of Currently Used Cell Culture Models**

Int J Mol Sci. 2023 Jun 20. >>[PubMed-Link](#)<<

Anna Rederer, Victoria Rose, René Krüger, Linda Schmittutz, Izabela Swierzy, Lena Fischer, Ingo Thievensen, Julian Bauer, Oliver Friedrich, Mario Schiffer, Janina Müller-Deile

#### 15. JUNI 2023 (CONNECT-GENERATE)

##### **Clinical associations and characteristics of the polyspecific intrathecal immune response in elderly patients with non-multiple sclerosis chronic autoimmune-inflammatory neurological diseases – a retrospective cross-sectional study**

Front Neurol. 2023 Jun 15. >>[PubMed-Link](#)<<

Felix Brauchle, Daniel Rapp, Makbule Senel, André Huss, Jens Dreyhaupt, Veronika Klose, Marie Süße, Klarissa Hanja Stürner, Frank Leypoldt, Hayrettin Tumani, Jan Lewerenz

#### 9. JUNI 2023 (CONNECT-GENERATE)

##### **Translational imaging of TSPO reveals pronounced innate inflammation in human and murine CD8 T cell-mediated limbic encephalitis**

Sci Adv. 2023 Jun 9. >>[PubMed-Link](#)<<

Marco Gallus, Wolfgang Roll, Andre Dik, Cristina Barca, Bastian Zinnhardt, Gordon Hicking, Christoph Mueller, Venu Narayanan Naik, Max Anstötz, Julia Krämer, Leoni Rolfes, Lydia Wachsmuth, Julika Pitsch, Karen M J van Loo, Saskia Räuber, Hideho Okada, Catriona Wimberley, Christine Strippel, Kristin S Golombeck, Andreas Johnen, Stjepana Kovac, Catharina C Groß, Philipp Backhaus, Robert Seifert, Jan Lewerenz, Rainer Surges, Christian E Elger, Heinz Wiendl, Tobias Ruck, Albert J Becker, Cornelius Faber, Andreas H Jacobs, Jan Bauer, Sven G Meuth, Michael Schäfers, Nico Melzer

#### 9. JUNI 2023 (TREAT-ION)

##### **KCNC2 variants of uncertain significance are also associated to various forms of epilepsy**

Front Neurol. 2023 Jun 9. >>[PubMed-Link](#)<<

Simone Seiffert, Manuela Pendziwiat, Ulrike B S Hedrich, Ingo Helbig, Yvonne Weber, Niklas Schwarz

#### 6. JUNI 2023 (GENERARE)

##### **Improvement of synaptic plasticity and cognitive function in RASopathies—a monocentre, randomized, double-blind, parallel-group, placebo-controlled, cross-over clinical trial (SynCoRAS)**

Trials. 2023 Jun 6. >>[PubMed-Link](#)<<

Nikolai H Jung, Silvia Egert-Schwender, Beate Schossow, Victoria Kehl, Ute Wahlländer, Louisa Brich, Viktoria Janke, Christiane Blankenstein, Martin Zenker, Volker Mall

#### 5. JUNI 2023 (GAIN)

##### **Management of a patient with common variable immunodeficiency and hepatopathy**

Allergy Asthma Clin Immunol. 2023 Jun 5. >>[PubMed-Link](#)<<

Lea Grümme, Hendrik Schulze-Koops

#### 2. JUNI 2023 (MITONET)

##### **Efficacy and Safety of Elamipretide in Individuals With Primary Mitochondrial Myopathy: The MMPOWER-3 Randomized Clinical Trial**

Neurology. 2023 Jun 2. >>[PubMed-Link](#)<<

Amel Karaa, Enrico Bertini, Valerio Carelli, Bruce H Cohen, Gregory M Enns, Marni J Falk, Amy Goldstein, Gráinne Siobhan 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, Russell Saneto, Fernando Scaglia, Serenella Servidei, Mark Tarnopolsky, Antonio Toscano, Johan L K Van Hove, John Vissing, Jerry Vockley, Jeffrey S Finman, David A Brown, James A Shiffer, Michelangelo Mancuso for the MMPOWER-3 Trial Investigators

#### 2. JUNI 2023 (STOP-FSGS)

##### **Characterizing Intraindividual Podocyte Morphology In Vitro with Different Innovative Microscopic and Spectroscopic Techniques**

Neurology. 2023 Jun 2. >>[PubMed-Link](#)<<

Annalena Kraus, Victoria Rose, René Krüger, George Sarau, Lasse Kling, Mario Schiffer, Silke Christiansen, Janina Müller-Deile

#### 1. JUNI 2023 (TREATHSP.NET)

##### **Heteromeric clusters of ubiquitinated ER-shaping proteins drive ER-phagy**

Nature. 2023 Jun. >>[PubMed-Link](#)<<

Hector Foronda, Yangxue Fu, Adriana Covarrubias-Pinto, Hartmut T Bocker, Alexis González, Eric Seemann, Patricia Franzka, Andrea Bock, Ramachandra M Bhaskara, Lutz Liebmann, Marina E Hoffmann, Istvan Katona, Nicole Koch, Joachim Weis, Ingo Kurth, Joseph G Gleeson, Fulvio Reggiori, Gerhard Hummer, Michael M Kessels, Britta Qualmann, Muriel Mari, Ivan Dikić, Christian A Hübner

#### 1. JUNI 2023 (STOP-FSGS)

##### **Accumulation of $\alpha$ -synuclein mediates podocyte injury in Fabry nephropathy**

J Clin Invest. 2023 Jun 1. >>[PubMed-Link](#)<<

Fabian Braun, Ahmed Abed, Dominik Sellung, Manuel Rogg, Mathias Woidy, Oysten Eikrem, Nicola Wanner, Jessica Gambardella, Sandra D Laufer, Fabian Haas, Milagros N Wong, Bernhard Dumoulin, Paula Rischke, Anne Mühligh, Wiebke Sachs, Katharina von Cossel, Kristina Schulz, Nicole Muschol, Sören W Gersting, Ania C Muntau, Oliver Kretz, Oliver Hahn, Markus M Rinschen, Michael Mauer, Tillmann Bork, Florian Grahammer, Wei Liang, Thorsten Eierhoff, Winfried Römer, Arne Hansen, Catherine Meyer-Schwesinger, Guido Iaccarino, Camilla Tøndel, Hans-Peter Marti, Behzad Najafian, Victor G Puelles, Christoph Schell, Tobias B Huber

#### 1. JUNI 2023 (ADDRESS)

##### **Willingness of individuals with Li-Fraumeni syndrome to participate in a cancer prevention trial: a survey study**

Fam Cancer. 2023 Jun 23. >>[PubMed-Link](#)<<

Farina J Struwe, Sarah Schott, Martina de Zwaan, Christian P Kratz

#### 1. JUNI 2023 (STOP-FSGS)

##### **Deep learning-based segmentation and quantification of podocyte foot process morphology suggests differential patterns of foot process effacement across kidney pathologies**

Kidney Int. 2023 Jun. >>[PubMed-Link](#)<<

David Unnersjö-Jess, Linus Butt, Martin Höhne, German Sergei, Arash Fatehi, Anna Witasp, Annika Wernerson, Jaakko Patrakka, Peter F Hoyer, Hans Blom, Bernhard Schermer, Katarzyna Bozek, Thomas Benzing

#### 1. JUNI 2023 (MITONET)

##### **Genotypic and phenotypic spectrum of infantile liver failure due to pathogenic TRMU variants**

Genet Med. 2023 Jun. >>[PubMed-Link](#)<<

Georg F Vogel, Yael Mozer-Glassberg, Yuval E Landau, Lea D Schlieben, Holger Prokisch, René G Feichtinger, Johannes A Mayr, Heiko Brennenstuhl, Julian Schröter, Agnes Pechlaner, Fowzan S Alkuraya, Joshua J Baker, Giulia Barcia, Ivo Baric, Nancy Braverman, Birute Burnyte, John Christodoulou, Elzbieta Ciara, David Coman,

Anibh M Das, Niklas Darin, Adela Della Marina, Felix Distelmaier, Erik A Eklund, Melike Ersoy, Weiyan Fang, Pauline Gaignard, Rebecca D Ganetzky, Emmanuel Gonzales, Caoimhe Howard, Joanne Hughes, Vassiliki Konstantopoulou, Melis Kose, Marina Kerr, Aneal Khan, Dominic Lenz, Robert McFarland, Merav Gil Margolis, Kevin Morrison, Thomas Müller, Kei Murayama, Emanuele Nicastro, Alessandra Pennisi, Heidi Peters, Dorota Piekutowska-Abramczuk, Agnès Rötig, René Santer, Fernando Scaglia, Manuel Schiff, Mohammad Shagrani, Mark Sharrard, Claudia Soler-Alfonso, Christian Staufner, Imogen Storey, Michael Stormon, Robert W Taylor, David R Thorburn, Elisa Leao Teles, Jian-She Wang, Daniel Weghuber, Saskia Wortmann

#### 29. MAI 2023 (STOP-FSGS)

##### **A previously uncharacterized Factor Associated with Metabolism and Energy (FAME/C14orf105/CCDC198/1700011H14Rik) is related to evolutionary adaptation, energy balance, and kidney physiology**

Nat Commun. 2023 May 29. >>[PubMed-Link](#)<<

Julian Petersen, Lukas Englmaier, Artem V Artemov, Irina Poverennaya, Ruba Mahmoud, Thibault Boudierlique, Marketa Tesarova, Ruslan Deviatiiarov, Anett Szilvásy-Szabó, Evgeny E Akkuratov, David Pajuelo Reguera, Hugo Zeberg, Marketa Kaucka, Maria Eleni Kastriti, Jan Krivanek, Tomasz Radaszkiewicz, Kristína Gömöryová, Sarah Knauth, David Potesil, Zbynek Zdrahal, Ranjani Sri Ganji, Anna Grabowski, Miriam E Buhl, Tomas Zikmund, Michaela Kavkova, Håkan Axelson, David Lindgren, Rafael Kramann, Christoph Kuppe, Ferenc Erdélyi, Zoltán Máté, Gábor Szabó, Till Koehne, Tibor Harkany, Kaj Fried, Jozef Kaiser, Peter Boor, Csaba Fekete, Jan Rozman, Petr Kaspárek, Jan Prochazka, Radislav Sedlacek, Vitezslav Bryja, Oleg Gusev, Igor Adameyko

#### **Published Erratum**

#### 27. MAI 2023 (TREATHSP.NET)

##### **Responsiveness of the SARA and natural history in 884 recessive and early onset ataxia patients**

Ann Neurol. 2023 May 27. >>[PubMed-Link](#)<<

Andreas Träschütz, Astrid D Adames-Gomez, Mathieu Anheim, Jonathan Baets, Bernard Brais, Cynthia Gagnon, Janina Gburek-Augustat, Sarah Doss, Hasmet A Hanagasi, Christoph Kamm, Peter Klivenyi, Thomas Klockgether, Thomas Klopstock, Martina Minnerop, Alexander Münchau, Mathilde Renaud, Filippo M Santorelli, Ludger Schöls, Andreas Thieme, Stefan Vielhaber, Bart P van de Warrenburg, Ginevra Zanni, Ralf-Dieter Hilgers, PREPARE consortium, Matthis Synofzik

#### 27. MAI 2023 (TREATHSP.NET)

##### **Responsiveness of the Scale for the Assessment and Rating of Ataxia and Natural History in 884 Recessive and Early Onset Ataxia Patients**

Ann Neurol. 2023 May 27. >>[PubMed-Link](#)<<

Andreas Träschütz, Astrid D Adames-Gómez, Mathieu Anheim, Jonathan Baets, Bernard Brais, Cynthia Gagnon, Janina Gburek-Augustat, Sarah Doss, Hasmet A Hanağasi, Christoph Kamm, Peter Klivenyi, Thomas Klockgether, Thomas Klopstock, Martina Minnerop, Alexander Münchau, Mathilde Renaud, Filippo M Santorelli, Ludger Schöls, Andreas Thieme, Stefan Vielhaber, Bart P van de Warrenburg, Ginevra Zanni, Ralf-Dieter Hilgers; PREPARE Consortium; Matthis Synofzik

#### 24. MAI 2023 (TREAT-ION)

##### **Loss or gain of function? Effects of ion channel mutations on neuronal firing depend on the neuron type**

Front Neurol. 2023 May 24. >>[PubMed-Link](#)<<

Nils A Koch, Lukas Sonnenberg, Ulrike B S Hedrich, Stephan Lauxmann, Jan Benda

#### 12. MAI 2023 (STOP-FSGS)

##### **Insights into human kidney function from the study of Drosophila**

Pediatr Nephrol. 2023 May 12. >>[PubMed-Link](#)<<

Sybille Koehler, Tobias B Huber

#### 9. MAI 2023 (MITONET)

##### **Age-dependent retinal neuroaxonal degeneration in children and adolescents with Leber hereditary optic neuropathy under idebenone therapy**

Eur J Neurol. 2023 May 9. >>[PubMed-Link](#)<<

Benedikt Schworm, Jakob Siedlecki, Claudia Catarino, Bettina von Livonius, Daniel R Muth, Guenther Rudolph, Joachim Havla, Thomas Klopstock, Claudia Priglinger

#### 9. MAI 2023 (HICHOL)

##### **Functional characterization of novel or yet uncharacterized ATP7B missense variants detected in patients with clinical Wilson's disease**

Clin Genet. 2023 May 9. >>[PubMed-Link](#)<<

Amelie Stalke, Annika Behrendt, Finja Hennig, Holger Gohlke, Nicole Buhl, Thea Reinkens, Ulrich Baumann, Brigitte Schlegelberger, Thomas Illig, Eva-Doreen Pfister, Britta Skawran

#### 8. MAI 2023 (TREATHSP.NET)

##### **Peripheral nerve involvement in hereditary spastic paraplegia characterized by quantitative magnetic resonance neurography**

Eur J Neurol. 2023 May 8. >>[PubMed-Link](#)<<

Heike Jacobi, Markus Weiler, Georges Sam, Sabine Heiland, John M Hayes, Martin Bendszus, Rebecca Schüle, Jennifer C Hayes

#### 4. MAI 2023 (GAIN)

##### **JAKs and STATs from a Clinical Perspective: Loss-of-Function Mutations, Gain-of-Function Mutations, and Their Multidimensional Consequences**

J Clin Immunol. 2023 May 4. >>[PubMed-Link](#)<<

Nils Ott, Laura Faletti, Maximilian Heeg, Virginia Andreani, Bodo Grimbacher

#### 4. MAI 2023 (TREAT-ION)

##### **CNV-ClinViewer: enhancing the clinical interpretation of large copy-number variants online**

Bioinformatics. 2023 May 4 >>[PubMed-Link](#)<<

Marie Macnee, Eduardo Pérez-Palma, Tobias Brünger, Chiara Klöckner, Konrad Platzer, Arthur Stefanski, Ludovica Montanucci, Allan Bayat, Maximilian Radtke, Ryan L Collins, Michael Talkowski, Daniel Blankenberg, Rikke S Møller, Johannes R Lemke, Michael Nothnagel, Patrick May, Dennis Lal

#### 3. MAI 2023 (CONNECT-GENERATE)

##### **Increased flexibility of brain dynamics in patients with multiple sclerosis**

Brain Commun. 2023 May 3. >>[PubMed-Link](#)<<

Nina von Schwanenflug, Stefan P Koch, Stephan Krohn, Tommy A A Broeders, David M Lydon-Staley, Dani S Bassett, Menno M Schoonheim, Friedemann Paul, Carsten Finke

#### 1. MAI 2023 (CONNECT-GENERATE)

##### **Immunoreactivity to astrocytes in different forms of dementia: High prevalence of autoantibodies to GFAP**

Brain Behav Immun Health. 2023 May. >>[PubMed-Link](#)<<

Paula Charlotte Barthel, Finja Staabs, Lucie Y Li, Maria Buthut, Carolin Otto, Klemens Ruprecht, Harald Prüss, Markus Höltje

#### 1. MAI 2023 (GAIN)

##### **Evaluation and Management of Deficiency of Adenosine Deaminase 2: An International Consensus Statement**

JAMA Netw Open. 2023 May 1. >>[PubMed-Link](#)<<

Pui Y Lee, Brad A Davidson, Roshini S Abraham, Blanche Alter, Juan I Arostegui, Katherine Bell, Alexandre Belot, Jenna R E Bergerson, Timothy J Bernard, Paul A Brogan, Yackov Berkun, Natalie T Deutch, Dimana Dimitrova, Sophie A GeorGIN-Lavialle, Marco Gattorno, Bodo Grimbacher, Hasan Hashem, Michael S Hershfield, Rebecca N Ichord, Kazushi Izawa, Jennifer A Kanakry, Raju P Khubchandani, Femke C C Klouwer, Evan A Luton, Ada W Man, Isabelle Meyts, Joris M Van Montfrans, Seza Ozen, Janna Saarela, Gustavo C Santo, Aman Sharma, Ariane Soldatos, Rachel Sparks, Troy R Torgerson, Ignacio Leandro Uriarte, Taryn A B Youngstein, Qing Zhou, Ivona Aksentijevich, Daniel L Kastner, Eugene P Chambers, Amanda K Ombrello; DADA2 Foundation

#### 1. MAI 2023 (GAIN)

##### **Humans with inherited MyD88 and IRAK-4 deficiencies are predisposed to hypoxemic COVID-19 pneumonia**

J Exp Med. 2023 May 1. >>[PubMed-Link](#)<<

Ana García-García, Rebeca Pérez de Diego, Carlos Flores, Darawan Rinchai, Jordi Solé-Violán, Àngela Deyà-Martínez, Blanca García-Solis, José M Lorenzo-Salazar, Elisa Hernández-Brito, Anna-Lisa Lanz, Leen Moens, Giorgia Bucciol, Mohamed Almuqamam, Joseph B Domachowske, Elena Colino, Juan Luis Santos-Perez, Francisco M Marco, Claudio Pignata, Aziz Bousfiha, Stuart E Turvey, Stefanie Bauer, Filomeen Haerynck, Javier Gonzalo Ocejo-Vinyals, Francisco Lendinez, Seraina Prader, Nora Naumann-Bartsch, Jana Pachlopnik Schmid, Catherine M Biggs, Kyla Hildebrand, Alexandra Dreesman, Miguel Ángel Cárdenes, Fatima Ailal, Ibtihal

Benhsaien, Giuliana Giardino, Agueda Molina-Fuentes, Claudia Fortuny, Swetha Madhavarapu, Daniel H Conway, Carolina Prando, Laire Schidlowski, María Teresa Martínez de Saavedra Álvarez, Rafael Alfaro, Felipe Rodríguez de Castro; ESID Registry Working Party; COVID Human Genetic Effort; Isabelle Meyts, Fabian Hauck, Anne Puel, Paul Bastard, Bertrand Boisson, Emmanuelle Jouanguy, Laurent Abel, Aurélie Cobat, Qian Zhang, Jean-Laurent Casanova, Laia Alsina, Carlos Rodríguez-Gallego

#### 1. MAI 2023 (MITONET)

##### **Aberrant splicing prediction across human tissues**

Nat Genet. 2023 May. >>[PubMed-Link](#)<<

Nils Wagner, Muhammed H Çelik, Florian R Hölzlwimmer, Christian Mertes, Holger Prokisch, Vicente A Yépez, Julien Gagneur

#### 1. MAI 2023 (STOP-FSGS)

##### **Down-regulation of human long non-coding RNA LINC01187 is associated with nephropathies**

J Cell Mol Med. 2023 May. >>[PubMed-Link](#)<<

Theodora Manolakou, Valeria Kaltezioti, Niki Prakoura, Panagiotis Kavadas, Simone Reichelt-Wurm, Harikleia Gakiopoulou, Miriam Banas, Bernhard Banas, Maja T Lindenmeyer, Clemens D Cohen, Peter Boor, Sonja Djudjaj, Dimitrios T Boumpas, Christos Chatziantoniou, Aristidis Charonis, Panagiotis K Politis

#### 28. APRIL 2023 (STOP-FSGS)

##### **Repetitive administration of rituximab can achieve and maintain clinical remission in patients with MCD or FSGS**

Sci Rep. 2023 Apr 28. >>[PubMed-Link](#)<<

Thomas Osterholt, Polina Todorova, Lucas Kühne, Rasmus Ehren, Lutz Thorsten Weber, Franziska Grundmann, Thomas Benzing, Paul Thomas Brinkkötter, Linus Alexander Völker

#### 21. APRIL 2023 (CONNECT-GENERATE)

##### **Preclinical safety and efficacy of a therapeutic antibody that targets SARS-CoV-2 at the sotrovimab face but is escaped by Omicron**

iScience. 2023 Apr 21. >>[PubMed-Link](#)<<

Jakob Kreye, S Momsen Reincke, Stefan Edelburg, Lara M Jeworowski, Hans-Christian Kornau, Jakob Trimpert, Peter Hombach, Sophia Halbe, Volker Nölle, Martin Meyer, Stefanie Kattenbach, Elisa Sánchez-Sendin, Marie L Schmidt, Tatjana Schwarz, Ruben Rose, Andi Krumbholz, Sophie Merz, Julia M Adler, Kathrin Eschke, Azza Abdelgawad, Dietmar Schmitz, Leif E Sander, Uwe Janssen, Victor M Corman, Harald Prüss

#### 21. APRIL 2023 (GAIN)

##### **The GAIN Registry – a New Prospective Study for Patients with Multi-organ Autoimmunity and Autoinflammation**

J Clin Immunol. 2023 Apr 21. >>[PubMed-Link](#)<<

Paulina Staus, Stephan Rusch, Sabine El-Helou, Gabriele Müller, Máté Krausz, Ulf Geisen, Andrés Caballero-Oteyza, Renate Krüger, Shahrzad Bakhtiar, Min Ae Lee-Kirsch, Maria Fasshauer, Ulrich Baumann, Bimba Franziska Hoyer, João Fabela Neves, Michael Borte, Maria Carrabba, Fabian Hauck, Stephan Ehl, Peter Bader, Horst von Bernuth, Faranaz Atschekzei, Mikko R J Seppänen, Klaus Warnatz, Alexandra Nieters, Gerhard Kindle, Bodo Grimbacher

#### 21. APRIL 2023 (HICHOL)

##### **Diagnostic workup of suspected hereditary cholestasis in adults: a case report**

Explor Dig Dis. 2023 Apr 21. >>[Link](#)<<

Carola Dröge, Tobias Götz, Annika Behrendt, Holger Gohlke, Verena Keitel

#### 17. APRIL 2023 (NEOCYST)

##### **Validation of attenuation imaging coefficient, shear wave elastography, and dispersion as emerging tools for non-invasive evaluation of liver tissue in children**

Front Pediatr. 2023 Apr 17. >>[PubMed-Link](#)<<

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

#### 13. APRIL 2023 (GENERARE)

##### **Editorial: Identifying the isoform-specific roles of RAS paralogs in health and disease**

Front Cell Dev Biol. 2023 Apr 13. >>[PubMed-Link](#)<<

Saeideh Nakhaei-Rad, Anna Fejtova

### 13. APRIL 2023 (NEOCYST)

#### **Clinical Characteristics and Courses of Patients With Autosomal Recessive Polycystic Kidney Disease-Mimicking Phenocopies**

Kidney Int Rep. 2023 Apr 13 >>[PubMed-Link](#)<<

Abdul A Halawi, Kathrin Burgmaier, Anja K Buescher, Ismail Dursun, Florian Erger, Matthias Galiano, Michaela Gessner, Ibrahim Gökce, Djalila Mekahli, Sevgi Mir, Lukasz Obrycki, Rukshana Shroff, Stella Stabouli, Maria Szczepanska, Ana Teixeira, Lutz T Weber, Andrea Wenzel Elke Wühl, Katarzyna Zachwieja, Jörg Dötsch, Franz Schaefer, Max C Liebau

### 10. APRIL 2023 (TREAT-ION)

#### **Expansion-enhanced super-resolution radial fluctuations enable nanoscale molecular profiling of pathology specimens**

Nat Nanotechnol. 2023 Apr >>[PubMed-Link](#)<<

Dominik Kylies, Marina Zimmermann, Fabian Haas, Maria Schwerk, Malte Kuehl, Michael Brehler, Jan Czogalla, Lola C Hernandez, Leonie Konczalla, Yusuke Okabayashi, Julia Menzel, Ilka Edenhofer, Sam Mezher, Hande Aypek, Bernhard Dumoulin, Hui Wu, Smilla Hofmann, Oliver Kretz, Nicola Wanner, Nicola M Tomas, Susanne Krasemann, Markus Glatzel, Christoph Kuppe, Rafael Kramann, Bella Banjanin, Rebekka K Schneider, Christopher Urbschat, Petra Arck, Nicola Gagliani, Marc van Zandvoort, Thorsten Wiech, Florian Grahammer, Pablo J Sáez, Milagros N Wong, Stefan Bonn, Tobias B Huber, Victor G Puelles

### 7. APRIL 2023 (CONNECT-GENERATE)

#### **Comparative Effects of Domain-Specific Human Monoclonal Antibodies Against LGI1 on Neuronal Excitability**

Neurol Neuroimmunol Neuroinflamm. 2023 Apr 7. >>[PubMed-Link](#)<<

Josefine Sell, Vahid Rahmati, Marin Kempfer, Sarosh R Irani, Andreas Ritzau-Jost, Stefan Hallermann, Christian Geis

### 7. APRIL 2023 (TREATHSP.NET)

#### **Autosomal Recessive Cerebellar Ataxias in Europe: Frequency, Onset, and Severity in 677 Patients**

Mov Disord. 2023 Apr 7. >>[PubMed-Link](#)<<

Andreas Träschütz, Astrid D Adarmes-Gomez, Mathieu Anheim, Jonathan Baets, Björn H Falkenburger, Janina Gburek-Augustat, Sarah Doss, Christoph Kamm, Peter Klivenyi, Marcus Grobe-Einsler, Thomas Klopstock, Martina Minnerop, Alexander Münchau, Chiara Pane, Mathilde Renaud, Filippo M Santorelli, Ludger Schöls, Dagmar Timmann, Stefan Vielhaber, Tobias B Haack, Bart P van de Warrenburg, Ginevra Zanni, Matthis Synofzik

### 7. APRIL 2023 (CONNECT-GENERATE)

#### **Microglia actively remove NR1 autoantibody-bound NMDA receptors and associated post-synaptic proteins in neuron microglia co-cultures**

Front Endocrinol (Lausanne). 2023 Mar 3. >>[PubMed-Link](#)<<

Kazi Atikur Rahman, Marta Orlando, Ayub Boulos 4, Ewa Andrzejak, Dietmar Schmitz, Noam E Ziv, Harald Prüss, Craig C Garner, Aleksandra Ichkova

### 1. APRIL 2023 (MYPRED)

#### **Rare and potentially fatal – Cytogenetically cryptic TNIP1::PDGFRB and PCM1::FGFR1 fusion leading to myeloid/lymphoid neoplasms with eosinophilia in children**

Cancer Genet. 2023 Apr. >>[PubMed-Link](#)<<

Ann-Cathrine Berking, Tim Flaadt, Yvonne Lisa Behrens, Ayami Yoshimi, Alfred Leipold, Ursula Holzer, Peter Lang, Leticia Quintanilla-Martinez, Brigitte Schlegelberger, Andreas Reiter, Charlotte Niemeyer, Brigitte Strahm, Gudrun Göhring

*joint publication ADDRESS/MyPred*

### 1. APRIL 2023 (ADDRESS)

#### **Rare and potentially fatal – Cytogenetically cryptic TNIP1::PDGFRB and PCM1::FGFR1 fusion leading to myeloid/lymphoid neoplasms with eosinophilia in children**

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Ann-Cathrine Berking, Tim Flaadt, Yvonne Lisa Behrens, Ayami Yoshimi, Alfred Leipold, Ursula Holzer, Peter Lang, Leticia Quintanilla-Martinez, Brigitte Schlegelberger, Andreas Reiter, Charlotte Niemeyer, Brigitte Strahm, Gudrun Göhring

*joint publication ADDRESS/MyPred*

### 1. APRIL 2023 (MYPRED)

#### **European standard clinical practice – Key issues for the medical care of individuals with familial leukemia**

Eur J Med Genet. 2023 Apr. >>[PubMed-Link](#)<<

Alisa Förster, Claudia Davenport, Nicolas Duployez, Miriam Erlacher, Alina Ferster, Jude Fitzgibbon, Gudrun Göhring, Henrik Hasle, Marjolijn C Jongmans, Alexandra Kolenova, Geertruijke Kronnie, Tim Lammens, Cristina Mecucci, Wojciech Mlynarski, Charlotte M Niemeyer, Francesc Sole, Tomasz Szczepanski, Esmé Waanders, Andrea Biondi, Marcin Wlodarski, Brigitte Schlegelberger, Tim Ripperger

*joint publication ADDRESS/MyPred*

### 1. APRIL 2023 (ADDRESS)

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*joint publication ADDRESS/MyPred*

### 1. APRIL 2023 (STOP-FSGS)

#### **Expansion-enhanced super-resolution radial fluctuations enable nanoscale molecular profiling of pathology specimens**

Nat Nanotechnol. 2023 Apr. >>[PubMed-Link](#)<<

Dominik Kylies, Marina Zimmermann, Fabian Haas, Maria Schwerk, Malte Kuehl, Michael Brehler, Jan Czogalla, Lola C Hernandez, Leonie Konczalla, Yusuke Okabayashi, Julia Menzel, Ilka Edenhofer, Sam Mezher, Hande Aypek, Bernhard Dumoulin, Hui Wu, Smilla Hofmann, Oliver Kretz, Nicola Wanner, Nicola M Tomas, Susanne Krasemann, Markus Glatzel, Christoph Kuppe, Rafael Kramann, Bella Banjanin, Rebekka K Schneider, Christopher Urbschat, Petra Arck, Nicola Gagliani, Marc van Zandvoort, Thorsten Wiech, Florian Grahammer, Pablo J Sáez, Milagros N Wong, Stefan Bonn, Tobias B Huber, Victor G Puelles

### 30. MÄRZ 2023 (HICHOL)

#### **Native liver survival in bile salt export pump deficiency: results of a retrospective cohort study**

Hepatol Commun. 2023 Mar 30. >>[PubMed-Link](#)<<

Eva-Doreen Pfister, Veronika K Jaeger, André Karch, Denys Shay, Nagoud Schukfeh, Johanna Ohlendorf, Norman Junge, Imeke Goldschmidt, Amelie Stalke, Verena Keitel-Anselmino, Ulrich Baumann

### 30. MÄRZ 2023 (GENERARE)

#### **Syndromic forms of congenital hyperinsulinism**

Front Endocrinol (Lausanne). 2023 Mar 3. >>[PubMed-Link](#)<<

Martin Zenker, Klaus Mohnike, Katja Palm

### 22. MÄRZ 2023 (CONNECT-GENERATE)

#### **Retrospective Pediatric Cohort Study Validates NEOS Score and Demonstrates Applicability in Children With Anti-NMDAR Encephalitis**

Neurol Neuroimmunol Neuroinflamm. 2023 Mar 22. >>[PubMed-Link](#)<<

Marc Nikolaus, Philipp Rausch, Kevin Rostásy, Annikki Bertolini, Ronny Wickström, Jessika Johannsen, Jonas Denecke, Markus Brey, Mareike Schimmel, Katharina Diepold, Martin Haeusler, Annegret Quade, Andrea Berger, Hendrik Rosewich, Claudia Steen, Katja von Au, Mona Dreesmann, Carsten Finke, Frederik Bartels, Angela M Kaindl, Markus Schuelke, Ellen Knierim

### 15. MÄRZ 2023 (GAIN)

#### **T-bet(high)CD21(low) B cells: the need to unify our understanding of a distinct B cell population in health and disease**

Curr Opin Immunol. 2023 Mar 15. >>[PubMed-Link](#)<<

Baerbel Keller, Klaus Warnatz

### 15. MÄRZ 2023 (TREATHSP.NET)

#### **Role of the adipocyte immune brain axis in Parkinson's disease: friend or foe?**

Neural Regen Res. 2023 Nov (online 2023 March 15) >>[PubMed-Link](#)<<

Martin Regensburger, Thomas M Kinfe



### 9. MÄRZ 2023 (MYPRED)

#### **RUNX1 isoform disequilibrium promotes the development of trisomy 21-associated myeloid leukemia**

Blood. 2023 Mar 9. >>[PubMed-Link](#)<<

Sofia Gialesaki, Daniela Bräuer-Hartmann, Hasan Issa, Raj Bhayadia, Oriol Alejo-Valle, Lonneke Verboon, Anna-Lena Schmell, Stephanie Laszig, Enikő Regényi, Konstantin Schuschel, Maurice Labuhn, Michelle Ng, Robert Winkler, Christian Ihling, Andrea Sinz, Markus Glaß, Stefan Hüttelmaier, Sören Matzk, Lena Schmid, Farina Josepha Strüwe, Sofie-Katrin Kadel, Dirk Reinhardt, Marie-Laure Yaspo, Dirk Heckl, Jan-Henning Klusmann

### 6. MÄRZ 2023 (TREAT-ION)

#### **Predicting functional effects of ion channel variants using new phenotypic machine learning methods**

PLoS Comput Biol. 2023 Mar 6. >>[PubMed-Link](#)<<

Christian Malte Boßelmann, Ulrike B S Hedrich, Holger Lerche, Nico Pfeifer

### 3. MÄRZ 2023 (TREATHSP.NET)

#### **Monogenetic Forms of Parkinson's Disease – Bridging the Gap Between Genetics and Biomarkers**

Front Aging Neurosci. 2022 Mar 3. >>[PubMed-Link](#)<<

Lars Tönges, Eun Hae Kwon, Stephan Klebe

### 1. MÄRZ 2023 (NEOCYST)

#### **A Low-Cost Sequencing Platform for Rapid Genotyping in ADPKD and its Impact on Clinical Care**

Kidney Int Rep. 2023 Mar. >>[PubMed-Link](#)<<

Christoph Heinrich Lindemann, Andrea Wenzel, Florian Erger, Lea Middelman, Julika Borde, Eric Hahnen, Denise Krauß, Simon Oehm, Sita Arjune, Polina Todorova, Kathrin Burgmaier, Max Christoph Liebau, Franziska Grundmann, Bodo B Beck, Roman-Ulrich Müller

### 1. MÄRZ 2023 (TREAT-ION)

#### **Generation of an induced pluripotent stem cell (iPSC) line from a patient with GEFS+ carrying a STX1B (p.Lys45delinsArgMetCysIleGlu and p.Leu46Met) mutation**

Stem Cell Res. 2023 Mar. >>[PubMed-Link](#)<<

Carolin Haag, Betül Uysal, Justus Marquetand, Heidi Löffler, Ulrike A Mau-Holzmann, Holger Lerche, Niklas Schwarz

### 1. MÄRZ 2023 (CONNECT-GENERATE)

#### **A genome-wide association study in autoimmune neurological syndromes with anti-GAD65 autoantibodies**

Brain. 2023 Mar 1 (published online 28.03.2022). >>[PubMed-Link](#)<<

Christine Strippel, Marisol Herrera-Rivero, Mareike Wendorff, ..., Nico Melzer; German Network for Research on Autoimmune Encephalitis (GENERATE)

**Podcast zur Publikation** im Podcast-Kanal des BRAIN-Journal (14.08.2023)

### 1. MÄRZ 2023 (STOP-FSGS)

#### **Precision nephrology: from molecular diagnostics to an individualized therapy**

Kidney Int. 2023 Mar. >>[PubMed-Link](#)<<

Anne K Mühlig, Jun Oh, Tobias B Huber

### 27. FEBRUAR 2023 (GAIN)

#### **Prospective Newborn Screening for SCID in Germany: A First Analysis by the Pediatric Immunology Working Group (API)**

J Clin Immunol. 2023 Feb 27. >>[PubMed-Link](#)<<

Carsten Speckmann, Uta Nennstiel, Manfred Hönig, Michael H Albert, Sujal Ghosh, Catharina Schuetz, Inken Brockow, Friederike Hörster, Tim Niehues, Stephan Ehl, Volker Wahn, Stephan Borte, Kai Lehmborg, Ulrich Baumann, Rita Beier, Renate Krüger, Shahrzad Bakhtiar, Joern-Sven Kuehl, Christian Klemann, Udo Kontny, Ursula Holzer, Andrea Meinhardt, Henner Morbach, Nora Naumann-Bartsch, Tobias Rothoefl, Alexandra Y Kreins, E Graham Davies, Dominik T Schneider, Horst V Bernuth, Thomas Klingebiel, Georg F Hoffmann, Ansgar Schulz, Fabian Hauck

### 24. FEBRUAR 2023 (MITONET)

#### **Genetics of mitochondrial diseases: Current approaches for the molecular diagnosis**

Handb Clin Neurol. 2023. >>[PubMed-Link](#)<<

Lea D Schlieben, Holger Prokisch

### 23. FEBRUAR 2023 (MYPRED)

#### **Second allogeneic stem cell transplantation can rescue a significant proportion of patients with JMML relapsing after first allograft**

Bone Marrow Transplant. 2023 Feb 23. >>[PubMed-Link](#)<<

Luca Vinci, Christian Flotho, Peter Noellke, Dirk Lebrecht, Riccardo Masetti, Valerie de Haas, Barbara De Moerloose, Michael Dworzak, Henrik Hasle, Tayfun Güngör, Jan Starý, Dominik Turkiewicz, Marek Ussowicz, Cristina Diaz de Heredia, Jochen Buechner, Kirsi Jahnukainen, Krisztian Kallay, Ivana Bodova, Owen P Smith, Marco Zecca, Dorine Bresters, Peter Lang, Tania Nicole Masmias, Roland Meisel, Herbert Pichler, Miriam Erlacher, Gudrun Göhring, Franco Locatelli, Brigitte Strahm, Charlotte M Niemeyer, Ayami Yoshimi

### 15. FEBRUAR 2023 (CONNECT-GENERATE)

#### **Glutamic acid decarboxylase antibody-associated neurological syndromes: Clinical and antibody characteristics and therapy response**

J Neurol Sci. 2023 Feb 15. >>[PubMed-Link](#)<<

Marie Madlener, Christine Strippel, Franziska S Thaler, Kathrin Doppler, Klaus P Wandinger, Jan Lewerenz, Marius Ringelstein, Rosa Roessling, Til Menge, Jonathan Wickel, Christoph Kellingshaus, Sigrid Mues, Andrea Kraft, Andreas Linsa, Simone C Tauber, Florian Then Berg, Stefan T Gerner, Asterios Paliantonis, Alexander Finke, Josef Priller, Ingo Schirotzek, Marie Süße, Kurt W Sühs, Christian Urbanek, Makbule Senel, Claudia Sommer, Tania Kuempfel, Harald Pruess, Gereon R Fink, Frank Leypoldt, Nico Melzer, Michael P Malter; German Network for Research on Autoimmune Encephalitis (GENERATE)

### 15. FEBRUAR 2023 (TREAT-ION)

#### **KCNA1 gain-of-function epileptic encephalopathy treated with 4-aminopyridine**

Ann Clin Transl Neurol. 2023 Feb 15. >>[PubMed-Link](#)<<

Peter Müller, Danielle S Takacs, Ulrike B S Hedrich, Rohini Coorg, Laura Masters, Kevin E Glinton, Hongzheng Dai, Jon A Cokley, James J Riviello, Holger Lerche, Edward C Cooper

### 13. FEBRUAR 2023 (TREAT-ION)

#### **Delineation of functionally essential protein regions for 242 neurodevelopmental genes**

Brain. 2023 Feb 13. >>[PubMed-Link](#)<<

Sumaiya Iqbal, Tobias Brünger, Eduardo Pérez-Palma, Marie Macnee, Andreas Brunklaus, Mark J Daly, Arthur J Campbell, David Hoksza, Patrick May, Dennis Lal

### 8. FEBRUAR 2023 (CONNECT-GENERATE)

#### **Impaired Brain Growth in Myelin Oligodendrocyte Glycoprotein Antibody-Associated Acute Disseminated Encephalomyelitis**

Neurol Neuroimmunol Neuroinflamm. 2023 Feb 8. >>[PubMed-Link](#)<<

Frederik Bartels, Birgit Baumgartner, Annette Aigner, Graham Cooper, Astrid Blaschek, Eva Maria Wendel, Annikki Bertolini, Michael Karenfort, Matthias Baumann, Robert Cleaveland, Andreas Wegener-Panzer, Steffen Leiz, Michela Salandin, Peter Krieg, Tobias Reindl, Markus Reindl, Carsten Finke, Kevin Rostás

### 6. FEBRUAR 2023 (GAIN)

#### **Human CARMIL2 deficiency underlies a broader immunological and clinical phenotype than CD28 deficiency**

J Exp Med. 2023 Feb 6. >>[PubMed-Link](#)<<

Romain Lévy, Florian Gothe, Mana Momenilandi, Thomas Magg, Marie Materna, Philipp Peters, Johannes Raedler, Quentin Philippot, Anita Lena Rack-Hoch, David Langlais, Mathieu Bourgey, Anna-Lisa Lanz, Masato Ogishi, Jérémie Rosain, Emmanuel Martin, Sylvain Latour, Natasha Vladikine, Marco Distefano, Taushif Khan, Franck Rapaport, Marian S Schulz, Ursula Holzer, Anders Fath, Georgios Sogkas, Carsten Speckmann, Arianna Troilo, Venetia Bigley, Anna Roppelt, Yael Dinur-Schejter, Ori Toker, Karen Helene Bronken Martinsen, Roya Sherkat, Ido Somekh, Raz Somech, Dror S Shouval, Jörn-Sven Kühl, Winnie Ip, Elizabeth M McDermott, Lucy Cliffe, Ahmet Ozen, Safa Baris, Hemalatha G Rangarajan, Emmanuelle Jouanguy, Anne Puel, Jacinta Bustamante, Marie-Alexandra Alyanakian, Mathieu Fusaro, Yi Wang, Xiao-Fei Kong, Aurélie Cobat, David Boutboul, Martin Castelle, Claire Aguilar, Olivier Hermine, Morgane Cheminant, Felipe Suarez, Alisan Yildiran, Aziz Bousfiha, Hamoud Al-Mousa, Fahad Alsohime, Deniz Cagdas, Roshini S Abraham, Alan P Knutsen, Borre Fevang, Sagar Bhattad, Ayca Kiykim, Baran Erman, Tugba Arikoglu, Ekrem Unal, Ashish Kumar, Christoph B Geier, Ulrich Baumann, Bénédicte Neven; CARMIL2 Consortium; Meino Rohlf, Christoph Walz, Laurent Abel, Bernard Malissen, Nico Marr, Christoph Klein, Jean-Laurent Casanova, Fabian Hauck, Vivien Béziat

#### 1. FEBRUAR 2023 (MITONET)

##### **Neuroimaging in mitochondrial disease**

Handb Clin Neurol. 2023. >>[PubMed-Link](#)<<  
Felix Distelmaier, Thomas Klopstock

#### 1. FEBRUAR 2023 (MITONET)

##### **Clinical trials in mitochondrial diseases**

Handb Clin Neurol. 2023. >>[PubMed-Link](#)<<  
Amel Karaa, Thomas Klopstock

#### 1. FEBRUAR 2023 (STOP-FSGS)

##### **The Role of Platelet-Derived Growth Factor in Focal Segmental Glomerulosclerosis**

J Am Soc Nephrol. 2023 Feb 1. >>[PubMed-Link](#)<<  
Ting Jia, Tong Xu, Bart Smeets, Eva Miriam Buhl, Marcus Johannes Moeller, Jürgen Floege, Barbara Mara Klinkhammer, Peter Boor

#### 1. FEBRUAR 2023 (CONNECT-GENERATE)

##### **Reduced resilience of brain state transitions in anti-N-methyl-D-aspartate receptor encephalitis**

Eur J Neurosci. 2023 Feb. >>[PubMed-Link](#)<<  
Nina von Schwanenflug, Juan P Ramirez-Mahaluf, Stephan Krohn, Amy Romanello, Josephine Heine, Harald Prüss, Nicolas A Crossley, Carsten Finke

#### 1. FEBRUAR 2023 (GENERARE)

##### **Genetic Variants in ARHGEF6 Cause Congenital Anomalies of the Kidneys and Urinary Tract in Humans, Mice, and Frogs**

J Am Soc Nephrol. 2023 Feb 1. >>[PubMed-Link](#)<<  
Verena Klämbt, Florian Buerger, Chunyan Wang, Thomas Naert, Karin Richter, Theresa Nauth, Anna-Carina Weiss, Tobias Sieckmann, Ethan Lai, Dervla M Connaughton, Steve Seltzsaam, Nina Mann, Amar J Majmundar, Chen-Han W Wu, Ana C Onuchic-Whitford, Shirlee Shril, Sophia Schneider, Luca Schierbaum, Rufeng Dai, Mir Reza Bekheirnia, Marieke Joosten, Omer Shlomovitz, Asaf Vivante, Ehud Banne, Shrikant Mane, Richard P Lifton, Karin M Kirschner, Andreas Kispert, Georg Rosenberger, Klaus-Dieter Fischer, Soeren S Lienkamp, Mirjam M P Zegers, Friedhelm Hildebrandt

#### 1. FEBRUAR 2023 (HICHOL)

##### **Cell-based BSEP trans-inhibition: A novel, non-invasive test for diagnosis of antibody-induced BSEP deficiency**

JHEP Rep. 2023 Feb 1. >>[PubMed-Link](#)<<  
Jan Stindt, Carola Dröge, Elke Lainka, Simone Kathemann, Eva-Doreen Pfister, Ulrich Baumann, Amelie Stalke, Enke Grabhorn, Mohammad Ali Shagrani, Yael Mozer-Glassberg, Jane Hartley, Marianne Wammers, Caroline Klindt, Paulina Philippski, Roman Liebe, Diran Herebian, Ertan Mayatepek, Thomas Berg, Anjona Schmidt-Choudhury, Constanze Wiek, Helmut Hanenberg, Tom Luedde, Verena Keitel

#### 30. JANUAR 2023 (TREAT-ION)

##### **Gentherapien für Epilepsie: Klinische Studien sind auf dem Weg [Gene Therapy for Epilepsy: Clinical Studies are on the Road]**

Fortschr Neurol Psychiatr. 2023 Jan 30. >>[PubMed-Link](#)<<  
Peter Müller, Holger Lerche

#### 28. JANUAR 2023 (STOP-FSGS)

##### **Next-Generation Morphometry for pathomics-data mining in histopathology**

Nat Commun. 2023 Jan 28. >>[PubMed-Link](#)<<  
David L Hölscher, Nassim Bouteldja, Mehdi Joodaki, Maria L Russo, Yu-Chia Lan, Alireza Vafaei Sadr, Mingbo Cheng, Vladimir Tesar, Saskia V Stillfried, Barbara M Klinkhammer, Jonathan Barratt, Jürgen Floege, Ian S D Roberts, Rosanna Coppo, Ivan G Costa, Roman D Bülow, Peter Boor

#### 24. JANUAR 2023 (GAIN)

##### **Common Variable Immunodeficiency: More Pathways than Roads to Rome**

Annu Rev Pathol. 2023 Jan 24. >>[PubMed-Link](#)<<  
Xiao P Peng, Andrés Caballero-Oteyza, Bodo Grimbacher

#### 19. JANUAR 2023 (CONNECT-GENERATE)

##### **Maternal synapsin autoantibodies are associated with neurodevelopmental delay**

Front Immunol. 2023 Jan 19. >>[PubMed-Link](#)<<

Isabel Bünger, Konstantin L Makridis, Jakob Kreye, Marc Nikolaus, Eva Sedlin, Tim Ullrich, Christian Hoffmann, Johannes Vincent Tromm, Helle Foverskov Rasmussen, Dragomir Milovanovic, Markus Höltje, Harald Prüss, Angela M Kaindl

#### 12. JANUAR 2023 (TREATHSP.NET)

##### **Deep Intronic FGF14 GAA Repeat Expansion in Late-Onset Cerebellar Ataxia**

N Engl J Med. 2023 Jan 12. >>[PubMed-Link](#)<<

David Pellerin, Matt C Danzi, Carlo Wilke, ..., Rebecca Schüle, Ludger Schöls, Roberta La Piana, Matthis Synofzik, Stephan Zuchner, Bernard Brais

#### 10. JANUAR 2023 (GAIN)

##### **Editorial: Updates on the pathogenesis of common variable immunodeficiency (CVID)**

Front Immunol. 2023 Jan 10. >>[PubMed-Link](#)<<

Timi Martelius, Mikko R J Seppänen, Klaus Warnatz

#### 10. JANUAR 2023 (STOP-FSGS)

##### **Single-cell transcriptomics reveals a mechanosensitive injury signaling pathway in early diabetic nephropathy**

Genome Med. 2023 Jan 10. >>[PubMed-Link](#)<<

Shuya Liu, Yu Zhao, Shun Lu, Tianran Zhang, Maja T Lindenmeyer, Viji Nair, Sydney E Gies, Guochao Wu, Robert G Nelson, Jan Czogalla, Hande Aypek, Stephanie Zielinski, Zhouning Liao, Melanie Schaper, Damian Fermin, Clemens D Cohen, Denis Delic, Christian F Krebs, Florian Grahmmer, Thorsten Wiech, Matthias Kretzler, Catherine Meyer-Schwesinger, Stefan Bonn, Tobias B Huber

#### 9. JANUAR 2023 (TREATHSP.NET)

##### **Mobile digital gait analysis objectively measures progression in hereditary spastic paraplegia**

Ann Clin Transl Neurol. 2023 Jan 9. >>[PubMed-Link](#)<<

Evelyn Loris, Malte Ollenschläger, Teresa Greinwalder, Björn Eskofier, Jürgen Winkler, Heiko Gaßner, Martin Regensburger

#### 6. JANUAR 2023 (CONNECT-GENERATE)

##### **Cerebrospinal fluid proteomics indicates immune dysregulation and neuronal dysfunction in antibody associated autoimmune encephalitis**

J Autoimmun. 2023 Jan 6. >>[PubMed-Link](#)<<

Saskia Räuber, Christina B Schroeter, Christine Strippel, Christopher Nelke, Tillmann Ruland, Andre Dik, Kristin S Golombeck, Liesa Regner-Nelke, Manuela Paunovic, Daniela Esser, Christian Münch, Felix Rosenow, Martijn van Duijn, Antonia Henes, Tobias Ruck, Ido Amit, Frank Leypoldt, Maarten J Titulaer, Heinz Wiendl, Sven G Meuth, Gerd Meyer Zu Hörste, Nico Melzer

#### 5. JANUAR 2023 (MITONET)

##### **Rare EIF4A2 variants are associated with a neurodevelopmental disorder characterized by intellectual disability, hypotonia, and epilepsy**

Am J Hum Genet. 2023 Jan 5. >>[PubMed-Link](#)<<

>>[Published Erratum](#)<<

Maimuna S Paul, Anna R Duncan, Casie A Genetti, Hongling Pan, Adam Jackson, Patricia E Grant, Jiahai Shi, Michele Pinelli, Nicola Brunetti-Pierrri, Alexandra Garza-Flores, Dave Shahani, Russell P Saneto, Giuseppe Zampino, Chiara Leoni, Emanuele Agolini, Antonio Novelli, Ulrike Blümlein, Tobias B Haack, Wolfram Heinritz, Eva Matzker, Bader Alhaddad, Rami Abou Jamra, Tobias Bartolomaeus, Saber AlHamdan, Raphael Carapito, Bertrand Isidor, Seiamak Bahram, Alyssa Ritter, Kosuke Izumi, Ben Pode Shakked, Ortal Barel, Bruria Ben Zeev, Amber Begtrup, Deanna Alexis Carere, Sureni V Mullegama, Timothy Blake Palculict, Daniel G Calame, Katharina Schwan, Alicia R P Aycinena, Rasa Traberg; Genomics England Research Consortium; Sofia Douzgou, Harrison Pirt, Naila Ismayilova, Siddharth Banka, Hsiao-Tuan Chao, Pankaj B Agrawal

2. JANUAR 2023 (GAIN)

**Inherited human ITK deficiency impairs IFN- $\gamma$  immunity and underlies tuberculosis**

J Exp Med. 2023 Jan 2. >>[PubMed-Link](#)<<

Masato Ogishi, Rui Yang, Rémy Rodriguez, Dominic P Golec, Emmanuel Martin, Quentin Philippot, Jonathan Bohlen, Simon J Pelham, Andrés Augusto Arias, Taushif Khan, Manar Ata, Fatima Al Ali, Flore Rozenberg, Xiao-Fei Kong, Maya Chrabieh, Candice Laine, Wei-Te Lei, Ji Eun Han, Yoann Seeleuthner, Zenia Kaul, Emmanuelle Jouanguy, Vivien Béziat, Leila Youssefian, Hassan Vahidnezhad, V Koneti Rao, Bénédicte Neven, Claire Fieschi, Davood Mansouri, Mohammad Shahrooei, Sevgi Pekcan, Gulsum Alkan, Melike Emiroğlu, Hüseyin Tokgöz, Jouni Uitto, Fabian Hauck, Jacinta Bustamante, Laurent Abel, Sevgi Keles, Nima Parvaneh, Nico Marr, Pamela L Schwartzberg, Sylvain Latour, Jean-Laurent Casanova, Stéphanie Boisson-Dupuis

1. JANUAR 2023 (TREAT-ION)

**Data-driven historical characterization of epilepsy-associated genes**

Eur J Paediatr Neurol. 2023 Jan. >>[PubMed-Link](#)<<

Marie Macnee, Eduardo Pérez-Palma, Javier A López-Rivera, Alina Ivaniuk, Patrick May, Rikke S Møller, Dennis Lal

1. JANUAR 2023 (CONNECT-GENERATE)

**Molecular disease mechanisms of human antineuronal monoclonal autoantibodies**

Trends Mol Med. 2023 Jan. >>[PubMed-Link](#)<<

Sophie L Duong, Harald Prüss

1. JANUAR 2023 (TREATHSP.NET)

**Emerging roles of leptin in Parkinson's disease: Chronic inflammation, neuroprotection and more?**

Brain Behav Immun. 2023 Jan. >>[PubMed-Link](#)<<

Martin Regensburger, Shafqat Rasul Chaudhry, Hammad Yasin, Yining Zhao, Andreas Stadlbauer, Michael Buchfelder, Thomas Kinfe