

## Publication list PID-NET

### **The NEW ESID online database network**

Bioinformatics 2019 Dec; 35(24): 5367-9

Scheible R, Rusch S, Guzman D, Mahlaoui N, Ehl S, Kindle G

1. OKTOBER 2019 (PID-NET)

### **DEUS: an R package for accurate small RNA profiling based on differential expression of unique sequences**

Bioinformatics 2019 Nov; 35(22): 4834-6

Jeske T, Huypens P, Stirn L, Hockele S, Wurmser CM, Bohm A, et al.

19. JULI 2019 (PID-NET)

### **The German National Registry of Primary Immunodeficiencies (2012-2017)**

Front. Immunol., 19 July 2019 | <https://doi.org/10.3389/fimmu.2019.01272>

Sabine M. El-Helou, et al.

1. JULI 2019 (PID-NET)

### **The German National Registry of Primary Immunodeficiencies (2012-2017)**

Front Immunol 2019 Jul; 10: 25

El-Helou SM, Biegner AK, Bode S, Ehl SR, Heeg M, Maccari ME, et al.

1. JUNI 2019 (PID-NET)

### **A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency**

J Allergy Clin Immunol 2019 Jun; 143(6): 2296-+

Volpi S, Cicalese MP, Tuijnenburg P, Tool ATJ, Cuadrado E, Abu-Halaweh M, et al.

22. MAI 2019 (PID-NET)

### **CARMIL2 Deficiency Presenting as Very Early Onset Inflammatory Bowel Disease**

Magg T, Shcherbina A, Arslan D, Desai MM, Wall S, Mitsialis V, Conca R, Unal E, Karacabey N, Mukhina A, Rodina Y, Taur PD, Illig D, Marquardt B, Hollizeck S, Jeske T, Gothe F, Schober T, Rohlf M, Koletzko S, Lurz E, Muise AM, Snapper SB, Hauck F, Klein C, Kotlarz D.

Inflamm Bowel Dis. 2019 May 22. pii: izz103. doi: 10.1093/ibd/izz103. [Epub ahead of print]

30. APRIL 2019 (PID-NET)

### **Preclinical Assessment of Suitable Natural Killer Cell Sources for Chimeric Antigen Receptor Natural Killer-Based „Off-the-Shelf“ Acute Myeloid Leukemia Immunotherapies**

Hum Gene Ther. 2019 Apr;30(4):381-401. doi: 10.1089/hum.2018.247. Epub 2019 Mar 18

Kloess S, Oberschmidt O, Dahlke J, Vu XK, Neudoerfl C, Kloos A, Gardlowski T, Matthies N, Heuser M, Meyer J, Sauer M, Falk C, Koehl U, Schambach A, Morgan MA.

21. MÄRZ 2019 (PID-NET)

### **Alternative Splicing Rescues Loss of Common Gamma Chain Function and Results in IL-21R-like Deficiency**

J Clin Immunol. 2019 Feb;39(2):207-215. doi: 10.1007/s10875-019-00606-7. Epub 2019 Mar 21. PMID: 30903457

Illig D, Navratil M, Kelečić J, Conca R, Hojsak I, Jadrašić O, Čorić M, Vuković J, Rohlf M, Hollizeck S, Bohne J, Klein C, Kotlarz D.

15. JANUAR 2019 (PID-NET)

### **Human RIPK1 deficiency causes combined immunodeficiency and inflammatory bowel diseases**

Proc Natl Acad Sci U S A. 2019 Jan 15;116(3):970-975. doi: 10.1073/pnas.1813582116.

Li Y, Führer M, Bahrami E, Socha P, Klaudel-Dreszler M, Bouzidi A, Liu Y, Lehle AS, Magg T, Hollizeck S, Rohlf M, Conca R, Field M, Warner N, Mordechai S, Shteyer E, Turner D, Boukari R, Belbouab R, Walz C, Gaidt MM, Hornung V, Baumann B, Pannicke U, Al Idrissi E, Ali Alghamdi H, Sepulveda FE, Gil M, de Saint Basile G, Hönig M, Koletzko S, Muise AM, Snapper SB, Schwarz K, Klein C, Kotlarz D.

1. JANUAR 2019 (PID-NET)

**Intestinal Inflammation and Dysregulated Immunity in Patients With Inherited Caspase-8 Deficiency**

Gastroenterology 2019 Jan; 156(1): 275-8

Lehle AS, Farin HF, Marquardt B, Michels BE, Magg T, Li Y, et al.

26. DEZEMBER 2018 (PID-NET)

**A patient-specific induced pluripotent stem cell model for West syndrome caused by ST3GAL3 deficiency**

Eur J Hum Genet. 2018 Dec;26(12):1773-1783. doi: 10.1038/s41431-018-0220-5. Epub 2018 Aug 8

van Diepen L, Buettner FFR, Hoffmann D, Thiesler CT, von Bohlen Und Halbach O, von Bohlen und Halbach V, Jensen LR, Steinemann D, Edvardson S, Elpeleg O, Schambach A, Gerardy-Schahn R, Kuss AW.

1. DEZEMBER 2018 (PID-NET)

**A patient-specific induced pluripotent stem cell model for West syndrome caused by ST3GAL3 deficiency**

Eur J Hum Genet 2018 Dec; 26(12): 1773-83

van Diepen L, Buettner FFR, Hoffmann D, Thiesler CT, Halbach OVU, Halbach VVU, et al.

29. OKTOBER 2018 (PID-NET)

**Engineering CAR-T Cells for Improved Function Against Solid Tumors**

Front Immunol. 2018 Oct 29;9:2493. doi: 10.3389/fimmu.2018.02493. eCollection 2018. Review

Morgan MA, Schambach A.

29. OKTOBER 2018 (PID-NET)

**Chimeric Antigen Receptor T Cells: Extending Translation from Liquid to Solid Tumors**

Hum Gene Ther. 2018 Oct;29(10):1083-1097. doi: 10.1089/hum.2017.251. Epub 2018 Aug 29

Morgan MA, Schambach A.

26. SEPTEMBER 2018 (PID-NET)

**Intestinal Inflammation and Dysregulated Immunity in Patients with Inherited Caspase-8 Deficiency**

Anna S. Lehle, Henner F. Farin, Benjamin Marquardt, Birgitta E. Michels, Thomas Magg, Yue Li, Yanshan Liu, Maryam Ghalandary, Katja Lammens, Sebastian Hollizeck, Meino Rohlf, Fabian Hauck, Raffaele Conca, Christoph Walz, Batia Weiss, Atar Lev, Amos J. Simon, Olaf Groß, Moritz M. Gaidt, Veit Hornung, Hans Clevers, Nadine Yazbeck, Rima Hanna-Wakim, Dror S. Shouval, Neil Warner, Raz Somech, Aleixo M. Muise, Scott S. Snapper, Philip Bufler, Sibylle Koletzko, Christoph Klein, Daniel Kotlarz

Gastroenterology. 2018 Sep 26. doi: 10.1053/j.gastro.2018.09.041. [Epub ahead of print]

DETAILS

1. JUNI 2018 (PID-NET)

**IPEX due to an exon 7 skipping FOXP3 mutation with autoimmune diabetes mellitus cured by selective T-Reg cell engraftment**

Clin Immunol 2018 Jun; 191: 52-8

Magg T, Wiebking V, Conca R, Krebs S, Arens S, Schmid I, et al.

23. APRIL 2018 (PID-NET)

**Rapid Flow Cytometry-Based Test for the Diagnosis of Lipopolysaccharide Responsive Beige-Like Anchor (LRBA) Deficiency**

Front Immunol. 2018 Apr 23;9:720. doi: 10.3389/fimmu.2018.00720. eCollection 2018.

Gámez-Díaz L, Sigmund EC, Reiser V, Vach W, Jung S, Grimbacher B.

1. MÄRZ 2018 (PID-NET)

**Recent advances in understanding the pathogenesis and management of reticular dysgenesis**

Br J Haematol 2018 Mar; 180(5): 644-53

Hoenig M, Pannicke U, Gaspar HB, Schwarz K

27. FEBRUAR 2018 (PID-NET)

**Human TGF- $\beta$ 1 deficiency causes severe inflammatory bowel disease and encephalopathy**

Nature Genetics 2018, published online 26 Februar 2018

Daniel Kotlarz, Benjamin Marquardt, Tuva Barøy, Way S. Lee, Liza Konnikova, Sebastian Hollizeck, Thomas Magg, Anna S. Lehle, Christoph Walz, Ingo Borggraefe, Fabian Hauck, Philip Bufler, Raffaele Conca, Sarah M. Wall, Eva M. Schumacher, Doriana Misceo, Eirik Frengen, Beint S. Bentsen, Holm H. Uhlig, Karl-Peter Hopfner, Aleixo M. Muise, Scott B. Snapper, Petter Strømme & Christoph Klein

DETAILS

15. NOVEMBER 2017 (PID-NET)

**Uncoupling the Oncogenic Engine**

Cancer Res. 2017 Nov 15;77(22):6060-6064. doi: 10.1158/0008-5472.CAN-17-2362. Epub 2017 Nov 2

Schambach A, Schott JW, Morgan MA.

31. OKTOBER 2017 (PID-NET)

**Integrating Vectors for Gene Therapy and Clonal Tracking of Engineered Hematopoiesis**

Hematol Oncol Clin North Am. 2017 Oct;31(5):737-752. doi: 10.1016/j.hoc.2017.06.009.

Biasco L, Rothe M, Schott JW, Schambach A.

21. OKTOBER 2017 (PID-NET)

**Myb-like, SWIRM, and MPN domains 1 (MYSM1) deficiency: Genotoxic stress-associated bone marrow failure and developmental aberrations**

J Allergy Clin Immunol. 2017 Oct;140(4):1112-1119. doi: 10.1016/j.jaci.2016.10.053.

Bahrami E, Witzel M, Racek T, Puchałka J, Hollizeck S, Greif-Kohistani N, Kotlarz D, Horny HP, Feederle R, Schmidt H, Sherkat R, Steinemann D, Göhring G, Schlegelbeger B, Albert MH, Al-Herz W, Klein C.

20. OKTOBER 2017 (PID-NET)

**Enhanced TH17 Responses in Patients with IL10 Receptor Deficiency and Infantile-onset IBD**

Inflamm Bowel Dis. 2017 Nov;23(11):1950-1961. doi: 10.1097/MIB.0000000000001270.

Shouval DS, Konnikova L, Griffith AE, Wall SM, Biswas A, Werner L, Nunberg M, Kammermeier J, Goettel JA, Anand R, Chen H, Weiss B, Li J, Loizides A, Yerushalmi B, Yanagi T, Beier R, Conklin LS, Ebens CL, Santos FGMS, Sherlock M, Goldsmith JD, Kotlarz D, Glover SC, Shah N, Bousvaros A, Uhlig HH, Muise AM, Klein C, Snapper SB

20. OKTOBER 2017 (PID-NET)

**MYSM1 deficiency- genotoxic stress-associated bone marrow failure and developmental aberrations**

J Allergy Clin Immunol. 2017 Oct;140(4):1112-1119. doi: 10.1016/j.jaci.2016.10.053.

Bahrami E, Witzel M, Racek T, Puchałka J, Hollizeck S, Greif-Kohistani N, Kotlarz D, Horny HP, Feederle R, Schmidt H, Sherkat R, Steinemann D, Göhring G, Schlegelbeger B, Albert MH, Al-Herz W, Klein C.

#### DETAILS

5. OKTOBER 2017 (PID-NET)

##### **Analyzing the Genotoxicity of Retroviral Vectors in Hematopoietic Cell Gene Therapy**

Mol Ther Methods Clin Dev. 2017 Oct 5;8:21-30. doi: 10.1016/j.omtm.2017.10.002. eCollection 2018 Mar 16. Review

Biasco L, Rothe M, Büning H, Schambach A.

5. OKTOBER 2017 (PID-NET)

##### **Analyzing the Genotoxicity of Retroviral Vectors in Hematopoietic Cell Gene Therapy**

Mol Ther Methods Clin Dev. 2017 Oct 5;8:21-30. doi: 10.1016/j.omtm.2017.10.002. eCollection 2018 Mar 16. Review

Biasco L, Rothe M, Büning H, Schambach A.

21. JULI 2017 (PID-NET)

##### **Multimodal Lentiviral Vectors for Pharmacologically Controlled Switching Between Constitutive Single Gene Expression and Tetracycline-Regulated Multiple Gene Collaboration.**

Hum Gene Ther Methods. 2017 Aug;28(4):191-204. doi: 10.1089/hgtb.2017.073. Epub 2017 Jul 5.

Stahlhut M, Schambach A, Kustikova OS.

21. JULI 2017 (PID-NET)

##### **Unique and shared signaling pathways cooperate to regulate the differentiation of human CD4+ T cells into distinct effector subsets.**

J Exp Med. 2016 Jul 25;213(8):1589-608. doi: 10.1084/jem.20151467. Epub 2016 Jul 11.

Ma CS, Wong N, Rao G, Nguyen A, Avery DT, Payne K, Torpy J, O'Young P, Deenick E, Bustamante J, Puel A, Okada S, Kobayashi M, Martinez-Barricarte R, Elliott M, Sebnem Kilic S, El Baghdadi J, Minegishi Y, Bousfiha A, Robertson N, Hambleton S, Arkwright PD, French M, Blincoe AK, Hsu P, Campbell DE, Stormon MO, Wong M, Adelstein S, Fulcher DA, Cook MC, Stepensky P, Boztug K, Beier R, Ikinçioğullari A, Ziegler JB, Gray P, Picard C, Boisson-Dupuis S, Phan TG, Grimbacher B, Warnatz K, Holland SM, Uzel G, Casanova JL, Tangye SG.

27. JUNI 2017 (PID-NET)

##### **Immunological phenotype of the murine Lrba knockout**

Immunol Cell Biol. 2017 Jun 27. doi: 10.1038/icb.2017.52

Gámez-Díaz L, Neumann J, Jäger F, Proietti M, Felber F, Soulas-Sprauel P, Perruzza L, Grassi F, Kögl T, Aichele P, Kilimann M, Grimbacher B, Jung S.

#### DETAILS

31. MAI 2017 (PID-NET)

##### **Potent and reversible lentiviral vector restriction in murine induced pluripotent stem cells**

Retrovirology. 2017 May 31;14(1):34. doi: 10.1186/s12977-017-0358-1.

Geis FK, Galla M, Hoffmann D, Kuehle J, Zychlinski D, Maetzig T, Schott JW, Schwarzer A, Goffinet C, Goff SP, Schambach A.

25. MAI 2017 (PID-NET)

##### **Chromatin-remodeling factor SMARCD2 regulates transcriptional networks controlling differentiation of neutrophil granulocytes**

Nat Genet. 2017 May;49(5):742-752. doi: 10.1038/ng.3833.

Witzel M, Petersheim D, Fan Y, Bahrami E, Racek T, Rohlf M, Puchařka J, Mertes C, Gagneur J, Ziegenhain C, Enard W, Stray-Pedersen A, Arkwright PD, Abboud MR, Pazhakh V, Lieschke GJ, Krawitz PM, Dahlhoff M, Schneider MR, Wolf E, Horny HP, Schmidt H, Schäffer AA, Klein C.

1. MAI 2017 (PID-NET)

**Reticular dysgenesis: international survey on clinical presentation, transplantation, and outcome**  
Blood 2017 May; 129(21): 2928-38

Hoenig M, Lagresle-Peyrou C, Pannicke U, Notarangelo LD, Porta F, Gennery AR, et al.

1. MAI 2017 (PID-NET)

**Clinical and Molecular Heterogeneity of RTEL1 Deficiency**  
Front Immunol 2017 May; 8: 19

Speckmann C, Sahoo SS, Rizzi M, Hirabayashi S, Karow A, Serwas NK, et al.

1. MAI 2017 (PID-NET)

**Clinical and Molecular Heterogeneity of RTEL1 Deficiency.**  
Front Immunol. 2017 May 1;8:449. doi: 10.3389/fimmu.2017.00449. eCollection 2017.

Speckmann C, Sahoo SS, Rizzi M, Hirabayashi S, Karow A, Serwas NK, Hoemberg M, Damatova N, Schindler D, Vannier JB, Boulton SJ, Pannicke U, Göhring G, Thomay K, Verdu-Amoros JJ, Hauch H, Woessmann W, Escherich G, Laack E, Rindle L, Seidl M, Rensing-Ehl A, Lausch E, Jandrasits C, Strahm B, Schwarz K, Ehl SR, Niemeyer C, Boztug K, Wlodarski MW.

24. APRIL 2017 (PID-NET)

**Detailed comparison of retroviral vectors and promoter configurations for stable and high transgene expression in human induced pluripotent stem cells.**  
Gene Ther. 2017 May;24(5):298-307. doi: 10.1038/gt.2017.20. Epub 2017 Apr 20.

Hoffmann D, Schott JW, Geis FK, Lange L, Muller FJ, Lenz D, Zychlinski D, Steinemann D, Morgan M, Moritz T, Schambach A.

16. FEBRUAR 2017 (PID-NET)

**Clinical spectrum and features of activated phosphoinositide 3-kinase  $\delta$  syndrome: A large patient cohort study.**  
J Allergy Clin Immunol. 2017 Feb;139(2):597-606.e4. doi: 10.1016/j.jaci.2016.06.021. Epub 2016 Jul 16.

Coulter TI, Chandra A, Bacon CM, Babar J, Curtis J, Screatton N, Goodlad JR, Farmer G, Steele CL, Leahy TR, Doffinger R, Baxendale H, Bernatoniene J, Edgar JD, Longhurst HJ, Ehl S, Speckmann C, Grimbacher B, Sediva A, Milota T, Faust SN, Williams AP, Hayman G, Kucuk ZY, Hague R, French P, Brooker R, Forsyth P, Herriot R, Cancrini C, Palma P, Ariganello P, Conlon N, Feighery C, Gavin PJ, Jones A, Imai K, Ibrahim MA, Markelj G, Abinun M, Rieux-Laucat F, Latour S, Pellier I, Fischer A, Touzot F, Casanova JL, Durandy A, Burns SO, Savic S, Kumararatne DS, Moshous D, Kracker S, Vanhaesebroeck B, Okkenhaug K, Picard C, Nejentsev S, Condliffe AM, Cant AJ

10. FEBRUAR 2017 (PID-NET)

**Kostmann's Disease and HCLS1-Associated Protein X-1 (HAX1)**  
J Clin Immunol. 2017 Feb;37(2):117-122. doi: 10.1007/s10875-016-0358-2.

Klein C.

1. FEBRUAR 2017 (PID-NET)

**Autoinhibition of the Nuclease ARTEMIS Is Mediated by a Physical Interaction between Its Catalytic and C-terminal Domains**  
J Biol Chem 2017 Feb; 292(8): 3351-65

Niewolik D, Peter I, Butscher C, Schwarz K

23. JANUAR 2017 (PID-NET)

**A human immunodeficiency syndrome caused by mutations in CARMIL2**

Nat Commun. 2017 Jan 23;8:14209

T. Schober, T. Magg, M. Laschinger, M. Rohlf, N. D. Linhares, J. Puchalka, T. Weisser, K. Fehlner, J. Mautner, C. Walz, K. Hussein, G. Jaeger, B. Kammer, I. Schmid, M. Bahia, S. D. Pena, U. Behrends, B. H. Belohradsky, C. Klein & F. Hauck

DETAILS

14. DEZEMBER 2016 (PID-NET)

**Gene Insertion Into Genomic Safe Harbors for Human Gene Therapy**

Mol Ther. 2016 Dec 14;24(4):678-84. doi: 10.1038/mt.2016.38. Review.

Papapetrou EP, Schambach A.

28. SEPTEMBER 2016 (PID-NET)

**XIAP deficiency and MEFV variants resulting in an autoinflammatory lymphoproliferative syndrome.**

BMJ Case Rep. 2016 Sep 28;2016. pii: bcr2016216922. doi: 10.1136/bcr-2016-216922

Christiansen M, Ammann S, Speckmann C, Mogensen TH

1. SEPTEMBER 2016 (PID-NET)

**Activated PI3K $\delta$  syndrome type 2: Two patients, a novel mutation, and review of the literature**

Pediatr Allergy Immunol. 2016 Sep;27(6):640-4

Olbrich P, Lorenz M, Cura Daball P, Lucena JM, Rensing-Ehl A, Sanchez B, Führer M, Camacho-Lovillo M, Melon M, Schwarz K, Neth O, Speckmann C

5. JULI 2016 (PID-NET)

**Viral and synthetic RNA vector technologies and applications**

Mol Ther. 2016 Jul 5. doi: 10.1038/mt.2016.143. [Epub ahead of print]

Wilhelmine Schott J, Morgan M, Galla M, Schambach A.

26. APRIL 2016 (PID-NET)

**Activated PI3K $\delta$  syndrome type 2: Two patients, a novel mutation and review of the literature**

Pediatr Allergy Immunol. 2016 Apr 26. doi: 10.1111/pai.12585. [Epub ahead of print]

Olbrich P, Lorenz M, Cura Daball P, Lucenas JM, Rensing-Ehl A, Sanchez B, Führer M, Camacho-Lovillo M, Melon M, Schwarz K, Neth O, Speckmann C.

26. APRIL 2016 (PID-NET)

**Alpharetroviral self-inactivating vectors produced by a superinfection-resistant stable packaging cell line allow genetic modification of primary human T lymphocytes**

Biomaterials. 2016 Aug;97:97-109. doi: 10.1016/j.biomaterials.2016.04.019. Epub 2016 Apr 26.

Labenski V, Suerth JD, Barczak E, Heckl D, Levy C, Bernadin O, Charpentier E, Williams DA, Fehse B, Verhoeven E, Schambach A.

24. APRIL 2016 (PID-NET)

**Functional Restoration of gp91phox-Oxidase Activity by BAC Transgenesis and Gene Targeting in X-linked Chronic Granulomatous Disease iPSCs**

Mol Ther 2015;24(4): 812-22.

Laugsch M, Rostovskaya M, Velychko S, Richter C, Zimmer A, Klink B, Schröck E, Haase M, Neumann K, Thieme S, Roesler J, Brenner S, Anastassiadis K.

14. APRIL 2016 (PID-NET)

**Hyperactive mTOR pathway promotes lymphoproliferation and abnormal differentiation in autoimmune lymphoproliferative syndrome**

Blood. 2016 Apr 20. pii: blood-2015-11-685024. [Epub ahead of print]

Völkl S, Rensing-Ehl A, Allgäuer A, Schreiner E, Lorenz MR, Rohr J, Klemann C, Fuchs I, Schuster V, von Bueren AO, Naumann-Bartsch N, Gambineri E, Siepermann K, Kobbe R, Nathrath M, Arkwright PD, Miano M, Stachel KD, Metzler M, Schwarz K, Kremer AN, Speckmann C, Ehl S, Mackensen A.

12. APRIL 2016 (PID-NET)

**Gene Insertion Into Genomic Safe Harbors for Human Gene Therapy.**

Mol Ther. 2016 Apr 12;24(4):678-84. doi: 10.1038/mt.2016.38. Epub 2016 Feb 12.

Papapetrou EP, Schambach A

1. APRIL 2016 (PID-NET)

**Caspase-1 regulates autoinflammation in rheumatic diseases**

Z Rheumatol. 2016; 75(3):265-75.

Winkler S, Hedrich CM, Rösen-Wolff A.

1. MÄRZ 2016 (PID-NET)

**Letter to the Editor: Production of Mature Healthy Hematopoietic Cells from Induced Pluripotent Stem Cells Derived from an AML Diagnostic Sample Containing the t(8;21) Translocation**

Stem Cells. 2016 Mar;34(3):797-9. doi: 10.1002/stem.2207.

Hoffmann D, Göhring G, Heuser M, Ganser A, Schambach A, Morgan MA.

23. FEBRUAR 2016 (PID-NET)

**Disturbed B-lymphocytes selection in autoimmune lymphoproliferative syndrome**

Blood. 2016 Feb 23. pii: blood-2015-04-642488. [Epub ahead of print]

Ales Janda, Klaus Schwarz, Mirjam van der Burg, Werner Vach, Hanna Ijspeert, Myriam Ricarda Lorenz, Magdeldin Elgizouli, Kathrin Pieper, Paul Fisch, Joachim Hagel, Raquel Lorenzetti, Maximilian Seidl, Joachim Roesler, Fabian Hauck, Elisabetta Traggiai, Carsten Speckmann, Anne Rensing-Ehl, Stephan Ehl, Hermann Eibel, Marta Rizzi

DETAILS

1. JANUAR 2016 (PID-NET)

**Efficient generation of gene-modified human natural killer cells via alpharetroviral vectors**

J Mol Med (Berl). 2016 Jan;94(1):83-93. doi: 10.1007/s00109-015-1327-6. Epub 2015 Aug 25.

Suerth JD, Morgan MA, Kloess S, Heckl D, Neudörfl C, Falk CS, Koehl U, Schambach A.

1. JANUAR 2016 (PID-NET)

**The extended phenotype of LPS-responsive beige-like anchor protein (LRBA) deficiency**

J Allergy Clin Immunol. 2016 Jan;137(1):223-30. doi: 10.1016/j.jaci.2015.09.025

Gámez-Díaz L, August D, Stepensky P, Revel-Vilk S, Seidel MG, Noriko M, Morio T, Worth AJ, Blessing J, Van de Veerdonk F, Feuchtinger T, Kanariou M, Schmitt-Graeff A, Jung S, Seneviratne S, Burns S, Belohradsky BH, Rezaei N, Bakhtiar S, Speckmann C, Jordan M, Grimbacher B

24. DEZEMBER 2015 (PID-NET)

**Neutrophil oxidative burst activates ATM to regulate cytokine production and apoptosis**

Blood. 2015;126(26): 2842-51.

Harbort CJ, Soeiro-Pereira PV, von Bernuth H, Kaindl AM, Costa-Carvalho BT, Condino-Neto A, Reichenbach J, Roesler J, Zychlinsky A, Amulic B.

20. DEZEMBER 2015 (PID-NET)

**DCLRE1C (ARTEMIS) mutations causing phenotypes ranging from atypical severe combined immunodeficiency to mere antibody deficiency**

Hum Mol Genet. 24, 7361-7372, 2015.

Volk, T., Pannicke, U., Reisli, I., Bulashevskaya, A., Ritter, J., Björkman, A., Schäffer, A.A., Fliegau, M., Sayar, E.H., Salzer, U., Fisch, P., Pfeifer, D., Di Virgilio, M., Cao, H., Yang, F., Zimmermann, K., Keles, S., Caliskaner, Z., Güner, S.Ü., Schindler, D., Hammarström, L., Rizzi, M., Hummel, M., Pan-Hammarström, Q., Schwarz, K. und Grimbacher, B.

1. DEZEMBER 2015 (PID-NET)

**SYK expression endows human ZAP70-deficient CD8 T cells with residual TCR signaling**

Clin Immunol. 161, 103-109, 2015.

Hauck, F., Blumenthal, B., Fuchs, S., Lenoir, C., Martin, E., Speckmann, C., Vraetz, T., Mannhardt-Laakmann, W., Lambert, N., Gil, M., Borte, S., Audrain, M., Schwarz, K., Lim, A., Schamel, W.W., Fischer, A., Ehl, S., Rensing-Ehl, A., Picard, C. und Latour, S.

1. NOVEMBER 2015 (PID-NET)

**TALEN-mediated functional correction of X-linked chronic granulomatous disease in patient-derived induced pluripotent stem cells**

Biomaterials. 2015 Nov;69:191-200. doi: 10.1016/j.biomaterials.2015.07.057.

Dreyer AK, Hoffmann D, Lachmann N, Ackermann M, Steinemann D, Timm B, Siler U, Reichenbach J, Grez M, Moritz T, Schambach A, Cathomen T.

24. OKTOBER 2015 (PID-NET)

**Retrovirus-based vectors for transient and permanent cell modification**

Curr Opin Pharmacol. 2015 Oct;24:135-46. doi: 10.1016/j.coph.2015.09.004. Epub 2015 Oct 1.

Schott JW, Hoffmann D, Schambach A.

15. OKTOBER 2015 (PID-NET)

**Gray platelet syndrome can mimic autoimmune lymphoproliferative syndrome**

Blood. 2015 Oct 15;126(16):1967-9. doi: 10.1182/blood-2015-06-654145

Rensing-Ehl A, Pannicke U, Zimmermann SY, Lorenz MR, Neven B, Fuchs I, Salzer U, Speckmann C, Strauss A, Maaß E, Collet B, Enders A, Favier R, Alessi MC, Rieux-Laucat F, Zieger B, Schwarz K, Ehl S

DETAILS

6. OKTOBER 2015 (PID-NET)

**Activating PI3Kδ mutations in a cohort of 669 patients with primary immunodeficiency**

Clin Exp Immunol. 2015 Oct 6. doi: 10.1111/cei.12706. [Epub ahead of print]

Elgizouli M\*, Lowe DM\*, Speckmann C\*, Schubert D, Hülsdünker J, Eskandarian Z, Dudek A, Schmitt-Graeff A, Wanders J, Jørgensen SF, Fevang B, Salzer U, Nieters A, Burns S, Grimbacher B. \*equal contribution

1. OKTOBER 2015 (PID-NET)

**Omenn syndrome associated with a functional reversion due to a somatic second-site mutation in CARD11 deficiency**

Blood 126, 1658-1669, 2015.

Fuchs, S.\*, Rensing-Ehl, A.\*, Pannicke, U.\*, Lorenz, M.R., Fisch, P., Jeelall, Y., Rohr, J., Speckmann, C., Vraetz, T., Farmand, S., Schmitt-Graeff, A., Krüger, M., Strahm, B., Henneke, P., Enders, A., Horikawa, K., Goodnow, C., Schwarz, K+ und Ehl,S+.

\* bzw. + equal contribution.

1. AUGUST 2015 (PID-NET)

**β2-Microglobulin deficiency causes a complex immunodeficiency of the innate and adaptive immune system**

J Allergy Clin Immunol.136, 392-401, 2015.

Ardeniz, Ö., Unger, S., Onay, H., Ammann, S., Keck, C., Cianga, C., Gerçeker, B., Martin, B., Fuchs, I.,



Salzer, U., İkinçioğulları, A., Güloğlu, D., Dereli, T., Thimme, R., Ehl, S., Schwarz, K., Schmitt-Graeff, A., Cianga, P., Fisch, P. und Warnatz, K.

1. JULI 2015 (PID-NET)

**Caspase-1: An integral regulator of innate immunity**

Semin Immunopathol. 2015 Jul;37(4):419-27. doi: 10.1007/s00281-015-0494-4. Epub 2015 Jun 10.

Winkler S and Rösen-Wolff A.

1. JULI 2015 (PID-NET)

**Current knowledge on procaspase-1 variants with reduced or abrogated enzymatic activity in autoinflammatory disease**

Curr Rheumatol Rep 17(7): 520-527

Luksch H, Winkler S, Heymann MC, Schulze F, Hofmann SR, Roesler J, and Rösen-Wolff A (2015)

1. JULI 2015 (PID-NET)

**Symptomatic males and female carriers in a large Caucasian kindred with XIAP deficiency**

J Clin Immunol. 2015 Jul;35(5):439-44. doi: 10.1007/s10875-015-0166-0. Epub 2015 May 6.

Dziadzio M, Ammann S, Canning C, Boyle F, Hassan A, Cale C, Elawad M, Fiil BK, Gyrd-Hansen M, Salzer U, Speckmann \*, Grimbacher B\*.

1. JULI 2015 (PID-NET)

**Mechanisms of clonal evolution in childhood acute lymphoblastic leukemia**

Nat Immunol. 16, 766-774, 2015.

Swaminathan, S., Klemm, L., Park, E., Papaemmanuil, E., Ford, A., Kweon, S.M., Trageser, D., Hasselfeld, B., Henke, N., Mooster, J., Geng, H., Schwarz, K., Kogan, S.C., Casellas, R., Schatz D,G., Lieber, M.R., Greaves, M.F. und Müschen, M.

1. JUNI 2015 (MD-NETPID-NET)

**Septic arthritis or juvenile idiopathic arthritis – the case of a 2 year old boy**

Pediatr Allergy Immunol. 26, 389-391, 2015.

Kallinich, T., Kölsch, U., Lieber, M., Unterwalder, N., Spors, B., Lorenz, M., Schwarz, K., Meisel, C. und von Bernuth, H.

27. MAI 2015 (PID-NET)

**Fluorescent tags influence the enzymatic activity and subcellular localization of procaspase-1**

Clin Immunol. 2015 May 27. pii: S1521-6616(15)00173-4. doi: 10.1016/j.clim.2015.05.011. [Epub ahead of print]

Heymann MC, Rabe S, Ruß S, Kapplusch F, Schulze F, Stein R, Winkler S, Hedrich CM, Rösen-Wolff A, Hofmann SR (2015)

15. MAI 2015 (PID-NET)

**Interleukin-10 receptor signaling in innate immune cells regulates mucosal immune tolerance and anti-inflammatory macrophage function**

Immunity 2014 May 15;40(5):706-19. Epub 2014 May 1

Shouval DS, Biswas A, Goettel JA, McCann K, Conaway E, Redhu NS, et al.

15. APRIL 2015 (PID-NET)

**CD4+ T Cell-Derived IL-21 and Deprivation of CD40 Signaling Favor the In Vivo Development of Granzyme B-Expressing Regulatory B Cells in HIV Patients**

J Immunol. 194, 3768-3777, 2015.

Kaltenmeier, C., Gawanbacht, A., Beyer, T., Lindner, S., Trzaska, T., van der Merwe, J.A., Härter, G., Grüner, B., Fabricius, D., Lotfi, R., Schwarz, K., Schütz, C., Hönig, M., Schulz, A., Kern, P., Bommer, M., Schrezenmeier, H., Kirchhoff, F. und Jahrsdörfer, B.

1. APRIL 2015 (PID-NET)

**Mutations in tetratricopeptide repeat domain 7A result in a severe form of very early onset inflammatory bowel disease**

Gastroenterology 2014 Apr;146(4):1028-39. doi: 10.1053/j.gastro.2014.01.015. Epub 2014 Jan 11.

Avitzur Y, Guo C, Mastropaolo LA, Bahrami E, Chen H, Zhao Z, et al...

1. APRIL 2015 (PID-NET)

**A Novel Thymoma-Associated Immunodeficiency with Increased Naive T Cells and Reduced CD247 Expression**

J Immunol. 194, 3045-3053, 2015.

Christopoulos, P., Dopfer, E.P., Malkovsky, M., Esser, P.R., Schaefer, H.E., Marx, A., Kock, S., Rupp, N., Lorenz, M.R., Schwarz, K., Harder, J., Martin, S.F., Werner, M., Bogdan, C., Schamel, W.W. und Fisch, P.

10. FEBRUAR 2015 (PID-NET)

**Large-scale hematopoietic differentiation of human induced pluripotent stem cells provides granulocytes or macrophages for cell replacement therapies**

Stem Cell Reports. 2015 Feb 10;4(2):282-96. doi: 10.1016/j.stemcr.2015.01.005

Lachmann N, Ackermann M, Frenzel E, Liebhaber S, Brenning S, Happle C, Hoffmann D, Klimenkova O, Lüttge D, Buchegger T, Kühnel MP, Schambach A, Janciauskiene S, Figueiredo C, Hansen G, Skokowa J, Moritz T.

8. FEBRUAR 2015 (PID-NET)

**Spectrum and management of complement immunodeficiencies (excluding hereditary angioedema) across Europe**

J Clin Immunol. 2015;35(2): 199-205.

Turley AJ, Gathmann B, Bangs C, Bradbury M, Seneviratne S, Gonzalez-Granado LI, Hackett S, Kutukculer N, Alachkar H, Hambleton S, Ritterbusch H, Kralickova P, Marodi L, Seidel MG, Dueckers G, Roesler J, Huissoon A, Baxendale H, Litzman J, Arkwright PD.

1. FEBRUAR 2015 (PID-NET)

**Infectious and immunologic phenotype of MECP2 duplication syndrome**

J Clin Immunol. 2015 Feb;35(2):168-81. doi: 10.1007/s10875-015-0129-5. Epub 2015 Feb 27.

Bauer M, Kölsch U, Krüger R, Unterwalder N, Hameister K, Kaiser FM, Vignoli A, Rossi R, Botella MP, Budisteanu M, Rosello M, Orellana C, Tejada MI, Papuc SM, Patat O, Julia S, Touraine R, Gomes T, Wenner K, Xu X, Afenjar A, Toutain A, Philip N, Jezela-Stanek A, Gortner L, Martinez F, Echenne B, Wahn V, Meisel C, Wiczorek D, El-Chehadeh S, Van Esch H, von Bernuth H.

29. JANUAR 2015 (PID-NET)

**Early-onset Evans syndrome, immunodeficiency, and premature immunosenescence associated with tripeptidyl-peptidase II deficiency**

Blood. 2015 Jan 29;125(5):753-61. doi: 10.1182/blood-2014-08-593202. Epub 2014 Nov 20

Stepensky P, Rensing-Ehl A, Gather R, Revel-Vilk S, Fischer U, Nabhani S, Beier F, Brümmendorf TH, Fuchs S, Zenke S, Firat E, Pessach VM, Borkhardt A, Rakhmanov M, Keller B, Warnatz K, Eibel H, Niedermann G, Elpeleg O, Ehl S.

1. DEZEMBER 2014 (PID-NET)

**The use of databases in primary immunodeficiencies**

Curr Opin Allergy Clin Immunol. 2014 Dec;14(6):501-8

Kindle G, Gathmann B, Grimbacher B

20. NOVEMBER 2014 (PID-NET)

**Early-onset Evans syndrome, immunodeficiency and premature immunosenescence associated with tripeptidyl-peptidase II deficiency**

Blood Epub

Stepensky P, Rensing-Ehl A, Gather R, et al.

21. OKTOBER 2014 (PID-NET)

**Combined immunodeficiency develops with age in immunodeficiency-centromeric instability facial anomalies syndrome (ICF2)**

Orphanet Journal of Rare Diseases 2014;9:113-118

von Bernuth H, Ravindran E, Du H, Fröhler S, Strehl K, Krämer N, Issa-Jahns L, Amulic B, Ninnemann O, Xiao MX, Erich K, Kölsch U, Hauptmann K, John R, Schindler D, Wahn V, Chen W, Kaindl AM

7. AUGUST 2014 (PID-NET)

**Improved retroviral episome transfer of transcription factors enables sustained cell fate modification**

Gene Ther. 2014 Nov;21(11):938-49. doi: 10.1038/gt.2014.69. Epub 2014 Aug 7.

Schott JW, Hoffmann D, Maetzig T, Müller FJ, Steinemann D, Zychlinski D, Cantz T, Baum C, Schambach A.

7. AUGUST 2014 (PID-NET)

**Abnormally differentiated CD4+ or CD8+ T cells with phenotypic and genetic features of double negative T cells in human Fas deficiency**

Blood. 2014 Aug 7;124(6):851-60. doi: 10.1182/blood-2014-03-564286. Epub 2014 Jun 3

Rensing-Ehl A, Völkl S, Speckmann C, Lorenz MR, Ritter J, Janda A, Abinun M, Pircher H, Bengsch B, Thimme R, Fuchs I, Ammann S, Allgäuer A, Kentouche K, Cant A, Hambleton S, Bettoni da Cunha C, Huetker S, Kühnle I, Pekrun A, Seidel MG, Hummel M, Mackensen A, Schwarz K, Ehl S.

1. JULI 2014 (PID-NET)

**Liver abscess complicated by diaphragm perforation and pleural empyema leads to the discovery of Interleukin-1 receptor associated kinase-4 (IRAK-4) deficiency**

Ped Infect Dis J. 2014 Jul. (Epub ahead of print)

Schöndorf D, von Bernuth H, Simon A, Schneider G, Kölsch U, Schwarz K, Meier CM, Große-Onnebring J, Gortner L, Rohrer TR

1. JULI 2014 (PID-NET)

**Clinical picture and treatment of 2212 patients with common variable immunodeficiency**

J Allergy Clin Immunol. 2014 Jul;134(1):116-26

Gathmann B, Mahlaoui N; CEREDIH, Gérard L, Oksenhendler E, Warnatz K, Schulze I, Kindle G, Kuijpers TW; Dutch WID, van Beem RT, Guzman D, Workman S, Soler-Palacín P, De Gracia J, Witte T, Schmidt RE, Litzman J, Hlavackova E, Thon V, Borte M, Borte S, Kumararatne D, Feighery C, Longhurst H, Helbert M, Szaflarska A, Sediva A, Belohradsky BH, Jones A, Baumann U, Meyts I, Kutukculer N, Wågström P, Galal NM, Roesler J, Farmaki E, Zinovieva N, Ciznar P, Papadopoulou-Alataki E, Bienemann K, Velbri S, Panahloo Z, Grimbacher B; European Society for Immunodeficiencies Registry Working Party

22. JUNI 2014 (PID-NET)

**Primary immunodeficiency diseases: an update on the classification from the international union of immunological societies expert committee for primary immunodeficiency**

Front Immunol. 2014;5:162

Al-Herz W, Bousfiha A, Casanova JL, Chatila T, Conley ME, Cunningham-Rundles C, et al.

12. JUNI 2014 (PID-NET)

**Inherited biallelic CSF3R mutations in severe congenital neutropenia**

2014 Jun 12; 123(24):3811-7. doi: 10.1182/blood-2013-11-535419. Epub 2014 Apr 21  
Triot A, Järvinen PM, Arostegui JI, Murugan D, Kohistani N, Dapena Díaz JL, et al..

1. JUNI 2014 (PID-NET)

**A new functional assay for the diagnosis of X-linked inhibitor of apoptosis (XIAP) deficiency**

Clin Exp Immunol. 2014 Jun;176(3):394-400. doi: 10.1111/cei.12306

Ammann S1, Elling R, Gyrd-Hansen M, Dückers G, Bredius R, Burns SO, Edgar JD, Worth A, Brandau H, Warnatz K, Zur Stadt U, Hasselblatt P, Schwarz K, Ehl S, Speckmann C.

2. MAI 2014 (PID-NET)

**JAGN1 deficiency causes aberrant myeloid cell homeostasis and congenital neutropenia**

Nat Genet.

Boztug K, Järvinen P, Salzer E, Racek T, Mönch S, Garncarz W, et al.

1. MAI 2014 (MD-NETPID-NET)

**Successful Haploidentical Hematopoietic Stem Cell Transplantation in a Patient with SCID due to CD3epsilon Deficiency: Need for IgG-Substitution 6 Years Later**

Klin Padiatr. 2014 May;226(3):149-53. Epub 2014 Feb 10.

Fuehrer M, Pannicke U, Schuetz C, Jacobsen EM, Schulz A, Friedrich W, et al.

1. MAI 2014 (PID-NET)

**Human procaspase-1 variants with decreased enzymatic activity are associated with febrile episodes and may contribute to inflammation via RIP2 and NF-κB signaling**

J Immunol. 2014 May 1;192(9):4379-85

Heymann MC, Wnkler S, Luksch H, Flecks S, Franke M, Ruß S, Ozen S, Yilmaz E, Klein C, Kallinich T, Lindemann D, Brenner S, Ganser G, Roesler J, Rösen-Wolff A, Hofmann SR

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22. APRIL 2014 (PID-NET)

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1. APRIL 2014 (PID-NET)

**Mutations in tetratricopeptide repeat domain 7A result in a severe form of very early onset inflammatory bowel disease**

Gastroenterology 2014 Apr;146(4):1028-39. doi: 10.1053/j.gastro.2014.01.015. Epub 2014 Jan 11.

Avitzur Y, Guo C, Mastropaolo LA, Bahrami E, Chen H, Zhao Z, et al...

1. APRIL 2014 (PID-NET)

**Lesson from hypomorphic recombination-activating gene (RAG) mutations: Why asymptomatic siblings should also be tested**

J Allergy Clin Immunol 2014 Apr;133(4):1211-5. Epub 2013 Dec 10.

Schuetz C., Pannicke U., Jacobsen E.-M., Burggraf S., Albert M.H., Hönig M., Niehues T., Feyen O., Ehl S., Debatin K.-M., Friedrich W., Schulz A.S. and Schwarz, K.

27. MÄRZ 2014 (PID-NET)

**Natural history of autoimmune lymphoproliferative syndrome associated with FAS gene mutations**

Blood. 2014 Mar 27;123(13):1989-99. Epub 2014 Jan 7

Price S, Shaw PA, Seitz A, Joshi G, Davis J, Niemela JE, et al.

12. MÄRZ 2014 (PID-NET)

**Gene Therapy for WiskottAldrich Syndrome—Long-Term Efficacy and Genotoxicity.**

Sci Transl Med. 2014 Mar 12;6(227):227ra33. doi: 10.1126/scitranslmed.3007280.

Braun CJ, Boztug K, Paruzynski A, Witzel M, Schwarzer A, Rothe M, Modlich U, Beier R, Göhring G, Steinemann D, Fronza R, Ball CR, Haemmerle R, Naundorf S, Kühlcke K, Rose M, Fraser C, Mathias L, Ferrari R, Abboud MR, Al-Herz W, Kondratenko I, Maródi L, Glimm H, Schlegelberger B, Schambach A, Albert MH, Schmidt M, von Kalle C, Klein C.

12. MÄRZ 2014 (PID-NET)

**Gene Therapy for Wiskott Aldrich Syndrome – Long-Term Efficacy and Genotoxicity**

Sci Transl Med. 2014 Mar 12;6(227):227ra33. doi: 10.1126/scitranslmed.3007280

Braun CJ, Boztug K, Paruzynski A, Witzel M, Schwarzer A, Rothe M, Modlich U, Beier R, Göhring G, Steinemann D, Fronza R, Ball CR, Haemmerle R, Naundorf S, Kühlcke K, Rose M, Fraser C, Mathias L, Ferrari R, Abboud MR, Al-Herz W, Kondratenko I, Maródi L, Glimm H, Schlegelberger B, Schambach A, Albert MH, Schmidt M, von Kalle C, Klein C.

12. FEBRUAR 2014 (PID-NET)

**Caspase-1 cleavage of the TLR adaptor TRIF inhibits autophagy and beta-interferon production during Pseudomonas aeruginosa infection**

Cell Host Microbe. 2014;Feb 12; 15(2):214-27

Jabir MS, Ritchie ND, Li D, Bayes HK, Tourlomousis P, Puleston D, Lupton A, Hopkins L, Simon AK, Bryant C, Evans TJ.

10. FEBRUAR 2014 (PID-NET)

**Successful haploidentical hematopoietic stem cell transplantation in a patient with SCID due to CD3ε deficiency**

Klin Padiatr. 2014 Feb 10. [Epub ahead of print] Fuehrer M., Pannicke U., Schuetz C., Schulz A., Friedrich W., Schwarz K. and Hoenig M.. (Contributors: Subproject A2).

17. JANUAR 2014 (PID-NET)

**Modified lentiviral LTRs allow Flp recombinase-mediated cassette exchange and in vivo tracing of “factor-free” induced pluripotent stem cells.**

Mol Ther. 2014 Jan 17.

Kuehle J, Turan S, Cantz T, Hoffmann D, Suerth JD, Maetzig T, Zychlinski D, Klein C, Steinemann D, Baum C, Bode J, Schambach A.

17. JANUAR 2014 (PID-NET)

**Modified lentiviral LTRs allow Flp recombinase-mediated cassette exchange and in vivo tracing of „factor-free“ induced pluripotent stem cells.**

Mol Ther. 2014 Jan 17.

Kuehle J, Turan S, Cantz T, Hoffmann D, Suerth JD, Maetzig T, Zychlinski D, Klein C, Steinemann D, Baum C, Bode J, Schambach A.

9. JANUAR 2014 (PID-NET)

**SCID patients with ARTEMIS vs RAG deficiencies following HCT: increased risk of late toxicity in ARTEMIS-deficient SCID**

Blood 2014 Jan 9; 123(2):281-9. Epub 2013 Oct 21.

Schuetz C., Neven B., Dvorak C.C., Leroy S., Ege M.J., Pannicke U., Schwarz, K., Schulz A.S., Honig, M., Sparber-Sauer, M., Gatz S.A., Denzer C., Blanche S., Moshous D., Picard C., Horn B.N., de Villartay J.P., Cavazzana M., Debatin K.M., Friedrich W., Fischer A. and Cowan M.J.

1. JANUAR 2014 (PID-NET)

**Dominant-activating germline mutations in the gene encoding the PI(3)K catalytic subunit p110delta result in T cell senescence and human immunodeficiency**

Nat Immunol. 2014 Jan;15(1):88-97. Epub 2013 Oct 28.

Lucas CL, Kuehn HS, Zhao F, Niemela JE, Deenick EK, Palendira U, et al.

1. JANUAR 2014 (PID-NET)

**Dominant-activating germline mutations in the gene encoding the PI(3)K catalytic subunit p110delta result in T cell senescence and human immunodeficiency**

Nat Immunol. 2014 Jan;15(1):88-97. Epub 2013 Oct 28.

Lucas CL, Kuehn HS, Zhao F, Niemela JE, Deenick EK, Palendira U, et al.

26. DEZEMBER 2013 (PID-NET)

**Deficiency of Innate and Acquired Immunity Caused by an IKBKB Mutation**

N Engl J Med. 2013 Dec 26;369(26):2504-14.

Pannicke, U., Baumann, B., Fuchs, S., Henneke, P., Rensing-Ehl, A., Rizzi, M., Janda, A., Hese, K., Schlesier, M., Holzmann, K., Borte, S., Laux, C., Rump, E.-M., Rosenberg, A., Zelinski, T., Schrezenmeier, H., Wirth, T., Ehl, S., Schroeder, M.L. und Schwarz, K.

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26. DEZEMBER 2013 (PID-NET)

**Mutations in AP3D1 associated with immunodeficiency and seizures define a new type of Hermansky-Pudlak syndrome**

N Engl J Med. 2013 Dec 26;369(26):2504-14. doi: 10.1056/NEJMoa1309199

Ammann S, Schulz A, Krägeloh-Mann I, Dieckmann NM, Niethammer K, Fuchs S, Eckl KM, Plank R, Werner R, Altmüller J, Thiele H, Nürnberg P, Bank J, Strauss A, von Bernuth H, Zur Stadt U, Grieve S, Griffiths GM, Lehmborg K, Hennies HC, Ehl S

26. DEZEMBER 2013 (PID-NET)

**Deficiency of Innate and Acquired Immunity Caused by an IKBKB Mutation**

N Engl J Med 2013; 369:2504-2514 December 26, 2013 DOI: 10.1056/NEJMoa1309199

Ulrich Pannicke, Ph.D., Bernd Baumann, Ph.D., Sebastian Fuchs, M.Sc., Philipp Henneke, M.D., Anne Rensing-Ehl, M.D., Marta Rizzi, M.D., Ph.D., Ales Janda, M.D., Ph.D., Katrin Hese, Ph.D., Michael Schlesier, Ph.D., Karlheinz Holzmann, Ph.D., Stephan Borte, M.D., Constanze Laux, Eva-Maria Rump, Alan Rosenberg, M.D., Teresa Zelinski, Ph.D., Hubert Schrezenmeier, M.D., Thomas Wirth, Ph.D., Stephan Ehl, M.D., Marlis L. Schroeder, M.D., and Klaus Schwarz, M.D.

9. DEZEMBER 2013 (PID-NET)

**Lesson from hypomorphic recombination-activating gene (RAG) mutations: Why asymptomatic siblings should also be tested.**

J Allergy Clin Immunol. 2013 Dec 9. pii: S0091-6749(13)01634-5

Schuetz, C., Pannicke, U., Jacobsen, E.-M., Burggraf, S., Albert, M.H., Hönig, M., Niehues, T., Feyen, O., Ehl, S., Debatin, K.-M., Friedrich, W., Schulz, A.S. und Schwarz, K.

1. DEZEMBER 2013 (PID-NET)

**Bromo- and extraterminal domain chromatin regulators serve as cofactors for murine leukemia virus integration.**

J Virol. 2013 Dec; 87(23):12721-36.

Gupta SS, Maetzig T, Maertens GN, Sharif A, Rothe M, Weidner-Glunde M, Galla M, Schambach A, Cherepanov P, Schulz TF.

1. DEZEMBER 2013 (PID-NET)

**Sequential decisions on FAS sequencing guided by biomarkers in patients with lymphoproliferation and autoimmune cytopenia.**

Haematologica. 2013 Dec;98(12):1948-55

Rensing-Ehl, A., Janda, A., Lorenz, M.R., Gladstone, B.P., Fuchs, I., Abinun, M., Albert, M., Butler, K., Cant, A., Cseh, A.M., Ebinger, M., Goldacker, S., Hambleton, S., Hebart, H., Houet, L., Kentouche, K.,

Kühnle, I., Lehmberg, K., Mejstrikova, E., Niemeyer, C., Minkov, M., Neth, O., Dückers, G., Owens, S., Rösler, J., Schilling, F.H., Schuster, V., Seidel, M.G., Smisek, P., Sukova, M., Svec, P., Wiesel, T., Gathmann, B., Schwarz, K., Vach, W., Ehl, S. und Speckmann, C.

26. NOVEMBER 2013 (PID-NET)

**Gene Correction of Human Induced Pluripotent Stem Cells Repairs the Cellular Phenotype in Pulmonary Alveolar Proteinosis.**

Am J Respir Crit Care Med. 2013 Nov 26. [Epub ahead of print]

Lachmann N, Happel C, Ackermann M, Lüttge D, Wetzke M, Merkert S, Hetzel M, Kensah G, Jara-Avaca M, Mucci A, Skuljec J, Dittrich AM, Pfaff N, Brenning S, Schambach A, Steinemann D, Göhring G, Cantz T, Martin U, Schwerk N, Hansen G, Moritz T.

26. NOVEMBER 2013 (PID-NET)

**Promoter and lineage independent anti-silencing activity of the A2 ubiquitous chromatin opening element for optimized human pluripotent stem cell-based gene therapy.**

Biomaterials. 2013 Nov 26.

Ackermann M, Lachmann N, Hartung S, Eggenschwiler R, Pfaff N, Happel C, Mucci A, Göhring G, Niemann H, Hansen G, Schambach A, Cantz T, Zweigerdt R, Moritz T.

6. NOVEMBER 2013 (PID-NET)

**Biosafety Challenges for Use of Lentiviral Vectors in Gene Therapy.**

Curr Gene Ther. 2013 Nov 6. [Epub ahead of print]

Rothe M, Modlich U, Schambach A.

1. NOVEMBER 2013 (PID-NET)

**Clinical Phenotype and Functional Analysis of a Rare XIAP/BIRC4 Mutation.**

Klin Pädiatr. 2013 Nov; 225(6):343-6

Vieth, S., Ammann, S., Schwarz, K., Härtel, C., Schultz, C., Lehmberg, K. und Lauten, M.

1. NOVEMBER 2013 (PID-NET)

**Gene therapy on the move.**

EMBO Mol Med. 2013 Nov; 5(11):1642-61.

Kaufmann KB, Büning H, Galy A, Schambach A, Grez M.

31. OKTOBER 2013 (PID-NET)

**High-resolution HLA matching in hematopoietic stem cell transplantation: a retrospective collaborative analysis.**

Blood. 2013 Oct 31;122(18):3220-9.

Fürst, D., Müller, C., Vucinic, V., Bunjes, D., Herr, W., Gramatzki, M., Schwerdtfeger, R., Arnold, R., Einsele, H., Wulf, G., Pfreundschuh, M., Glass, B., Schrezenmeier, H., Schwarz, K. und Mytilineos, J.

21. OKTOBER 2013 (PID-NET)

**SCID patients with ARTEMIS deficiency need myeloablation for full immune reconstitution following allogeneic HCT but have increased risk for late toxicity compared to patients with RAG deficient SCID.**

Blood. 2013 Oct 21. [Epub ahead of print]

Schuetz, C., Neven, B., Dvorak, C.C., Horn, B., Leroy, S., Ege, M., Pannicke, U., Schwarz, K., Schulz, A.S., Honig, M., Sparber-Sauer, M., Gatz, S.A., Denzer, C., Blanche, S., Moshous, D., Picard, C., de Villartay, J.P., Cavazzana-Calvo, M., Debatin, K.M., Friedrich, W., Fischer, A. und Cowan, M.J.

3. OKTOBER 2013 (PID-NET)

**Successful treatment of an infant with CDA type II by intrauterine transfusions and postnatal stem cell transplantation.**

Pediatr Blood Cancer 2013 Oct 3.

Braun; M., Wölfel, M., Wiegering, V., Winkler, B., Ertan, K., Bald, R., Schwarz, K., Heimpel, H., Eyrich, M. und Schlegel, P.G.

1. OKTOBER 2013 (PID-NET)

**X-linked inhibitor of apoptosis (XIAP) deficiency: The spectrum of presenting manifestations beyond hemophagocytic lymphohistiocytosis.**

Clin Immunol. 2013 Oct;149(1):133-41

Speckmann, C., Lehmborg, K., Albert, M.H., Damgaard, R.B., Fritsch, M., Gyrd-Hansen, M., Rensing-Ehl, A., Vraetz, T., Grimbacher, B., Salzer, U., Fuchs, I., Ufheil, H., Belohradsky, B.H., Hassan, A., Cale, C.M., Elawad, M., Strahm, B., Schibli, S., Lauten, M., Kohl, M., Meerpohl, J.J., Rodeck, B., Kolb, R., Eberl, W., Soerensen, J., von Bernuth, H., Lorenz, M., Schwarz, K., Zur Stadt, U. und Ehl, S.

1. SEPTEMBER 2013 (PID-NET)

**Even in Pneumococcal Sepsis CD62L Shedding on Granulocytes Proves to be a Reliable Functional Test for the Diagnosis of Interleukin-1 Receptor Associated Kinase 4 (IRAK-4) Deficiency**

Ped Infect Dis J 2013; 32(9):1017-9 (\*equal contribution)

Strehl K\*, Andres O\*, Kölsch U, Kunzmann S, Lebrun AH, Stroh T, Schwarz K, Liese J\*, von Bernuth H\*

1. SEPTEMBER 2013 (PID-NET)

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