

Publikationen des Verbundes 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- β 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 δ 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 δ 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 δ 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

DETAILS

22. APRIL 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.

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.

DETAILS

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., Lehmborg, K., Mejstrikova, E., Niemeyer, C., Minkov, M., Neth, O., Dücker, 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., Lehmborg, 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)

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 deficiency.

Pediatr Infect Dis J. 2013 Sep;32(9):1017-9.

Andres, O., Strehl, K., Kölsch, U., Kunzmann, S., Lebrun, A.H., Stroh, T., Schwarz, K., Morbach, H., von Bernuth, H. und Liese, J.

1. AUGUST 2013 (PID-NET)

The German national registry for primary immunodeficiencies (PID).

Clin Exp Immunol. 2013 Aug;173(2):372-80. doi: 10.1111/cei.12105.

Gathmann B, Goldacker S, Klima M, Belohradsky BH, Notheis G, Ehl S, Ritterbusch H, Baumann U, Meyer-Bahlburg A, Witte T, Schmidt R, Borte M, Borte S, Linde R, Schubert R, Bienemann K, Laws HJ, Dueckers G, Roesler J, Rothoefl T, Krüger R, Scharbatke EC, Masjosthusmann K, Wasmuth JC, Moser O, Kaiser P, Groß-Wieltsch U, Classen CF, Horneff G, Reiser V, Binder N, El-Helou SM, Klein C, Grimbacher B, Kindle G.

19. JULI 2013 (PID-NET)

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

Haematologica. 2013 Jul 19. [Epub ahead of print]

Rensing-Ehl A, Janda A, Lorenz MR, Gladstone BP, Fuchs I, Abinun M, Albert M, Butler K, Cant AJ, Cseh AM, Ebinger M, Goldacker S, Hambleton S, Hebart H, Houet L, Kentouche K, Kühnle I, Lehmborg K, Mejstrikova E, Niemeyer C, Minkov M, Neth O, Dücker G, Owens S, Roesler J, Schilling FH, Schuster V, Seidel MG, Smisek P, Sukova M, Svec P, Wiesel T, Gathmann B, Schwarz K, Vach W, Ehl S, Speckmann C.

4. JULI 2013 (PID-NET)

A congenital neutrophil defect syndrome associated with mutations in VPS45.

N Engl J Med. 2013 Jul 4;369(1):54-65. doi: 10.1056/NEJMoa1301296. Epub 2013 Jun 5.

Vilboux T, Lev A, Malicdan MC, Simon AJ, Järvinen P, Racek T, Puchalka J, Sood R, Carrington B, Bishop K, Mullikin J, Huizing M, Garty BZ, Eyal E, Wolach B, Gavrieli R, Toren A, Soudack M, Atawneh OM, Babushkin T, Schiby G, Cullinane A, Avivi C, Polak-Charcon S, Barshack I, Amariglio N, Rechavi G, van der Werff ten Bosch J, Anikster Y, Klein C, Gahl WA, Somech R.

1. JULI 2013 (PID-NET)

Therapeutic management of primary immunodeficiency in older patients.

Drugs Aging. 2013 Jul;30(7):503-12. doi: 10.1007/s40266-013-0079-7.

Verma N, Thaventhiran A, Gathmann B; ESID Registry Working Party, Thaventhiran J, Grimbacher B.

1. JULI 2013 (PID-NET)

Alu repeat-induced deletions in chronic granulomatous disease: a cause not only for p67-phox, but also for p47-phox deficiency.

Ann Hematol. 2013 Jul;92(7):1003-4. doi: 10.1007/s00277-012-1661-5. Epub 2012 Dec 29.

Winkler S, van Leeuwen K, Deboer M, Rösen-Wolff A, Roos D, Roesler J.

1. MAI 2013 (PID-NET)

Contribution of the inflammasomes to autoinflammatory diseases and recent mouse models as research tools.

Clin Immunol. 2013 Jun;147(3):175-84. doi: 10.1016/j.clim.2013.01.006. Epub 2013 Feb 4.

Heymann MC, Rösen-Wolff A.

15. APRIL 2013 (PID-NET)

Outcomes of splenectomy in patients with common variable immunodeficiency (CVID): a survey of 45 patients.

Clin Exp Immunol. 2013 Apr;172(1):63-72. doi: 10.1111/cei.12039.

Wong GK, Goldacker S, Winterhalter C, Grimbacher B, Chapel H, Lucas M, Alecsandru D, McEwen D, Quinti I, Martini H, Schmidt RE, Ernst D, Espanol T, Vidaller A, Carbone J, Fernandez-Cruz E, Lougaris V, Plebani A, Kutukculer N, Gonzalez-Granado LI, Contreras R, Kiani-Alikhan S, Ibrahim MA, Litzman J, Jones A, Gaspar HB, Hammarstrom L, Baumann U, Warnatz K, Huissoon AP; Clinical Working Party of the European Society for Immunodeficiencies (ESID).

11. APRIL 2013 (PID-NET)

The risk of hemophagocytic lymphohistiocytosis in Hermansky-Pudlak syndrome type 2.

Blood. 2013 Apr 11;121(15):2943-51. doi: 10.1182/blood-2012-10-463166. Epub 2013 Feb 12.

Jessen B, Bode SF, Ammann S, Chakravorty S, Davies G, Diestelhorst J, Frei-Jones M, Gahl WA, Gochuico BR, Griese M, Griffiths G, Janka G, Klein C, Kögl T, Kurnik K, Lehmborg K, Maul-Pavicic A, Mumford AD, Pace D, Parvaneh N, Rezaei N, de Saint Basile G, Schmitt-Graeff A, Schwarz K, Karasu GT, Zieger B, Zur Stadt U, Aichele P, Ehl S.

8. APRIL 2013 (PID-NET)

Somatic loss of heterozygosity, but not haploinsufficiency alone, leads to full-blown autoimmune lymphoproliferative syndrome in 1 of 12 family members with FAS start codon mutation.

Clin Immunol. 2013 Apr;147(1):61-8. doi: 10.1016/j.clim.2013.02.019. Epub 2013 Mar 5

Hauck F, Magerus-Chatinet A, Vicca S, Rensing-Ehl A, Roesen-Wolff A, Roesler J, Rieux-Laucat F.

1. APRIL 2013 (PID-NET)

Inflammatory Bowel Disease and T cell Lymphopenia in G6PC3 Deficiency.

J Clin Immunol. 2013 Apr;33(3):520-5. doi: 10.1007/s10875-012-9833-6. Epub 2012 Nov 20.

Begin P, Patey N, Mueller P, Rasquin A, Sirard A, Klein C, Haddad E, Drouin E, Le Deist F.

11. MÄRZ 2013 (PID-NET)

Loss-of-function mutations in the IL-21 receptor gene cause a primary immunodeficiency syndrome.

J Exp Med. 2013 Mar 11;210(3):433-43. doi: 10.1084/jem.20111229. Epub 2013 Feb 25.

Kotlarz D, Zietara N, Uzel G, Weidemann T, Braun CJ, Diestelhorst J, Krawitz PM, Robinson PN, Hecht J, Puchalka J, Gertz EM, Schäffer AA, Lawrence MG, Kardava L, Pfeifer D, Baumann U, Pfister ED, Hanson EP, Schambach A, Jacobs R, Kreipe H, Moir S, Milner JD, Schwille P, Mundlos S, Klein C.

1. MÄRZ 2013 (PID-NET)

A ubiquitous chromatin opening element prevents transgene silencing in pluripotent stem cells and their differentiated progeny.

Stem Cells. 2013 Mar;31(3):488-99.

Pfaff N, Lachmann N, Ackermann M, Kohlscheen S, Brendel C, Maetzig T, Niemann H, Antoniou MN, Grez M, Schambach A, Cantz T, Moritz T.

15. FEBRUAR 2013 (PID-NET)

Risk factors and prognosis in T-cell posttransplantation lymphoproliferative diseases: reevaluation of 163 cases.

Transplantation. 2013 Feb 15;95(3):479-88. doi: 10.1097/TP.0b013e3182762e07.

Tiede C, Maecker-Kolhoff B, Klein C, Kreipe H, Hussein K.

2. FEBRUAR 2013 (PID-NET)

Biosafety features of lentiviral vectors.

Hum Gene Ther. 2013 Feb;24(2):132-42. doi: 10.1089/hum.2012.229.

Schambach A, Zychlinski D, Ehrnstroem B, Baum C.

1. FEBRUAR 2013 (PID-NET)

Small amyloidogenic HIV-1 gp120 fragments boost retro-and lentiviral gene transfer.

Nat Nanotechnol. 2013 Feb;8(2):130-6.

Yolamanova, M., Arnold, F., Zirafi, O., Müller, J., Sauter, D., Goffinet, C., Reisser, M., Vas, V., Geiger, H., Lunov, O., Simmet, T., Bohne, J., Eisele, K., Meier, C., Weil, T., Schrezenmeier, H., Schwarz, K., Kirchhoff, F. und Münch, J.

1. FEBRUAR 2013 (PID-NET)

Animal models of human granulocyte diseases.

Hematol Oncol Clin North Am. 2013 Feb;27(1):129-48, ix. doi: 10.1016/j.hoc.2012.10.005. Epub 2012 Oct 31.

Schäffer AA, Klein C.

1. FEBRUAR 2013 (PID-NET)

Preface. Neutropenia.

Hematol Oncol Clin North Am. 2013 Feb;27(1):xi-xii. doi: 10.1016/j.hoc.2012.11.005. Epub 2012 Nov 27.

Klein C.

1. FEBRUAR 2013 (PID-NET)

Genetics and pathophysiology of severe congenital neutropenia syndromes unrelated to neutrophil elastase.

Hematol Oncol Clin North Am. 2013 Feb;27(1):43-60, vii. doi: 10.1016/j.hoc.2012.11.004. Epub 2012 Nov 27.

Boztug K, Klein C.

4. JANUAR 2013 (PID-NET)

Naturally occurring genetic variants of human caspase-1 differ considerably in structure and the ability to activate interleukin-1 β .

Hum Mutat. 2013 Jan;34(1):122-31. doi: 10.1002/humu.22169. Epub 2012 Aug 10.

Luksch H, Romanowski MJ, Chara O, Tüngler V, Caffarena ER, Heymann MC, Lohse P, Aksentijevich I, Remmers EF, Flecks S, Quoos N, Gramatté J, Petzold C, Hofmann SR, Winkler S, Pessler F, Kallinich T, Ganser G, Nimtz-Talaska A, Baumann U, Runde V, Grimbacher B, Birmelin J, Gahr M, Roesler J, Rösen-Wolff A.

1. JANUAR 2013 (PID-NET)

IL-10R polymorphisms are associated with very-early-onset ulcerative colitis.

Inflamm Bowel Dis. 2013 Jan;19(1):115-23. doi: 10.1002/ibd.22974.

Moran CJ, Walters TD, Guo CH, Kugathasan S, Klein C, Turner D, Wolters VM, Bandsma RH, Mouzaki M, Zachos M, Langer JC, Cutz E, Benseler SM, Roifman CM, Silverberg MS, Griffiths AM, Snapper SB, Muise AM.

1. JANUAR 2013 (PID-NET)

Characteristics of early and late PTLD development in pediatric solid organ transplant recipients.

Transplantation. 2013 Jan 15;95(1):240-6. doi: 10.1097/TP.0b013e318277e344.

Schober T, Framke T, Kreipe H, Schulz TF, Großhennig A, Hussein K, Baumann U, Pape L, Schubert S, Wingen AM, Jack T, Koch A, Klein C, Maecker-Kolhoff B.

17. OKTOBER 2012 (PID-NET)

A complex immunodeficiency is based on U1 snRNP-mediated poly(A) site suppression.

EMBO J. 2012 Oct 17;31(20):4035-44. doi: 10.1038/emboj.2012.252. Epub 2012 Sep 11.

Langemeier J, Schrom EM, Rabner A, Radtke M, Zychlinski D, Saborowski A, Bohn G, Mandel-Gutfreund Y, Bodem J, Klein C, Bohne J.

1. OKTOBER 2012 (PID-NET)

Attenuated TLR4/MAPK signaling in monocytes from patients with CRMO results in impaired IL-10 expression.

Clin Immunol. 2012 Oct;145(1):69-76. doi: 10.1016/j.clim.2012.07.012. Epub 2012 Aug 4.

Hofmann SR, Morbach H, Schwarz T, Rösen-Wolff A, Girschick HJ, Hedrich CM.

8. SEPTEMBER 2012 (PID-NET)

Profiling of the human monocytic cell secretome by quantitative label-free mass spectrometry identifies stimulus-specific cytokines and proinflammatory proteins.

Proteomics. 2012 Sep;12(18):2833-42. doi: 10.1002/pmic.201200108.

Groessl M, Luksch H, Rösen-Wolff A, Shevchenko A, Gentzel M.

1. SEPTEMBER 2012 (PID-NET)

“Mutation negative“ Familial Cold Autoinflammatory Syndrome (FCAS) in an 8-year-old boy – Clinical course and functional studies.

Rheumatol Internat 32: 2629-36.

Hedrich CM, Bruck N, Paul D, Hahn G, Gahr M, Roesen-Wolff A

1. SEPTEMBER 2012 (PID-NET)

Coexistence of sickle cell disease and severe congenital neutropenia: first impressions can be deceiving.

Eur J Haematol. 2012 Sep;89(3):245-9. doi: 10.1111/j.1600-0609.2012.01827.x.

Wali Y, Beshlawi I, Fawaz N, Alkhayat A, Zalabany M, Elshinawy M, Al-Kindi S, Al-Rawas AH, Klein C.

1. AUGUST 2012 (PID-NET)

Loss of Interleukin-10 Signaling and Infantile Inflammatory Bowel Disease: Implications for Diagnosis and Therapy.

Gastroenterology. 2012 Aug;143(2):347-55. doi: 10.1053/j.gastro.2012.04.045. Epub 2012 Apr 28.

Kotlarz D, Beier R, Murugan D, Diestelhorst J, Jensen O, Boztug K, Pfeifer D, Kreipe H, Pfister ED, Baumann U, Puchalka J, Bohne J, Egritas O, Dalgic B, Kolho KL, Sauerbrey A, Buderus S, Gungor T, Enninger A, Koda YK, Guariso G, Weiss B, Corbacioglu S, Socha P, Uslu N, Metin A, Wahbeh GT, Husain K, Ramadan D, Al-Herz W, Grimbacher B, Sauer M, Sykora KW, Koletzko S, Klein C.

8. JULI 2012 (PID-NET)

Clinical outcome in IL-10- and IL-10 receptor-deficient patients with or without hematopoietic stem cell transplantation.

The Journal of allergy and clinical immunology, 2012

Engelhardt KR, Shah N, Faizura-Yeop I, Kocacik Uygun DF, Frede N, Muise AM, Shteyer E, Filiz S, Chee R, Elawad M, Hartmann B, Arkwright PD, Dvorak C, Klein C, Puck JM, Grimbacher B, Glocker EO.

8. JULI 2012 (PID-NET)

Schwerer kombinierter Immundefekt.

In: Klinische Immunologie. Eds.: H.-H. Peter, W.J. Pichler und U. Müller-Ladner Elsevier, 190-199, 2012.

Schwarz, K. und Hönig, M.

1. JULI 2012 (PID-NET)

Delayed-onset adenosine deaminase deficiency: Strategies for an early diagnosis.

J Allergy Clin Immunol. 130, 991-994, 2012.

Speckmann, C., Neumann, C., Borte, S., La Marca, G., Sass, J.O., Wiech, E., Fisch, P., Schwarz, K., Buchholz, B., Schlesier, M., Felgentreff, K., Grimbacher, B., Santisteban, I., Bali, P., Hershfield, M.S. und Ehl, S.

8. JUNI 2012 (PID-NET)

Challenges in vector and trial design using retroviral vectors for long-term gene correction in hematopoietic stem cell gene therapy.

Mol Ther. 2012 Jun;20(6):1084-94. doi: 10.1038/mt.2012.93. PubMed PMID: 22652996; PubMed Central

Corrigan-Curay J, Cohen-Haguenauer O, O'Reilly M, Ross SR, Fan H, Rosenberg N, Somia N, King N, Friedmann T, Dunbar C, Aiuti A, Naldini L, Baum C, von Kalle C, Kiem HP, Montini E, Bushman F, Sorrentino BP, Carrondo M, Malech H, Gahrton G, Shapiro R, Wolff L, Rosenthal E, Jambou R, Zaia J, Kohn DB.

1. JUNI 2012 (PID-NET)

CXCL13 as a Novel Marker for Diagnosis and Disease Monitoring in Pediatric PTLD.

Am J Transplant. 2012 Jun;12(6):1610-7. doi: 10.1111/j.1600-6143.2011.03968.x. Epub 2012 Feb 15.

Schiffer L, Henke-Gendo C, Wilsdorf N, Hussein K, Pape L, Schmitt C, Haller H, Schiffer M, Klein C, Kreipe H, Maecker-Kolhoff B.

8. MAI 2012 (PID-NET)

Alpharetroviral self-inactivating vectors: long-term transgene expression in murine hematopoietic cells and low genotoxicity.

Mol Ther. 2012 May;20(5):1022-32. doi: 10.1038/mt.2011.309. Epub 2012 Feb 14.

Suerth JD, Maetzig T, Brugman MH, Heinz N, Appelt JU, Kaufmann KB, Schmidt M, Grez M, Modlich U, Baum C, Schambach A.

1. MAI 2012 (PID-NET)

Induced transgene expression for the treatment of solid tumors by hematopoietic stem cell-based gene therapy.

Cancer Gene Ther. 2012 May;19(5):352-7. doi: 10.1038/cgt.2012.8. Epub 2012 Mar 9.

Noyan F, Diez IA, Hapke M, Klein C, Dewey RA.

12. APRIL 2012 (PID-NET)

The phenotype of human STK4 deficiency.

Blood. 2012;119(15):3450-7.

Abdollahpour H, Appaswamy G, Kotlarz D, Diestelhorst J, Beier R, Schaeffer AA, Gertz EM, Schambach A, Kreipe HH, Pfeifer D, Engelhardt KR, Rezaei N, Grimbacher B, Lohrmann S, Sherkat R, Klein C.

1. APRIL 2012 (PID-NET)

Extended Spectrum of Human Glucose-6-Phosphatase Catalytic Subunit 3 Deficiency: Novel Genotypes and Phenotypic Variability in Severe Congenital Neutropenia.

J Pediatr. 2012 Apr;160(4):679-683.e2. doi: 10.1016/j.jpeds.2011.09.019. Epub 2011 Nov 1.

Boztug K, Rosenberg PS, Dorda M, Banka S, Moulton T, Curtin J, Rezaei N, Corns J, Innis JW, Avci Z, Hung Chi T, Pellier I, Pierani P, Fruge R, Parvaneh N, Mamishi S, Mody R, Darbyshire P, Motwani J, Murray J, Buchanan GR, Newman WG, Alter BP, Boxer LA, Donadieu J, Welte K, Klein C.

1. MÄRZ 2012 (PID-NET)

Allogeneic hematopoietic SCT for alpha-mannosidosis: an analysis of 17 patients.

Bone Marrow Transplant. 2012 Mar;47(3):352-9. doi: 10.1038/bmt.2011.99. Epub 2011 May 9.

Mynarek M, Tolar J, Albert MH, Escolar ML, Boelens JJ, Cowan MJ, Finnegan N, Glomstein A, Jacobsohn DA, Kuhl JS, Yabe H, Kurtzberg J, Malm D, Orchard PJ, Klein C, Lucke T, Sykora KW.

1. FEBRUAR 2012 (PID-NET)

Immunological reconstitution in a patient with ZAP-70 deficiency following transfusion of blood lymphocytes from a previously transplanted sibling without conditioning.

Bone Marrow Transplantation 47, 305–307, 2012.

Hönig, M., Schuetz, C., Schwarz, K., Rojewski, M., Jacobsen, E., Lahr, G., Debatin, K.M., Schulz, A. und Friedrich, W.

1. FEBRUAR 2012 (PID-NET)

Antiviral and regulatory T cell immunity in a patient with stromal interaction molecule 1 deficiency.

J Immunol. 188, 1523-1533, 2012.

Fuchs, S., Rensing-Ehl, A., Speckmann, C., Bengsch, B., Schmitt-Gräff, A., Maul-Pavicic, A., Vraetz, T., Strahm, B., Ankermann, T., Caliebe, A., Fölster-Holst, R., Kaiser, P., Thimme, R., Schwarz, K., Feske, S. und Ehl, S.

1. DEZEMBER 2011 (PID-NET)

IL-10 and IL-10 receptor defects in humans.

Year in Human and Medical Genetics: Inborn Errors of Immunity

Ann N Y Acad Sci. 2011 Dec;1246:102-7. doi: 10.1111/j.1749-6632.2011.06339.x.

Glocker E-O, Kotlarz D, Klein C, Shah N, Grimbacher B, Annals NYAS.

1. DEZEMBER 2011 (PID-NET)

Chronic non-bacterial osteomyelitis is associated with impaired Sp1 signaling, reduced IL10 promoter phosphorylation, and reduced myeloid IL-10 expression.

Clin Immunol 141: 317-327.

Hofmann SR, Schwarz T, Möller JC, Morbach H, Schnabel A, Rösen-Wolff A, Girschick HJ, Hedrich CM

1. DEZEMBER 2011 (PID-NET)

Gene therapy for SCID-X1: focus on clinical data.

Mol Ther. 2011

Dec;19(12):2103-4. doi: 10.1038/mt.2011.252. PubMed PMID: 22134740; PubMed Central

Baum C.

1. NOVEMBER 2011 (PID-NET)

Elevated serum BAFF levels in patients with autoimmunity and lymphoproliferation.

Scand J Immunol. 2011 Nov;74(5):518-9. doi: 10.1111/j.1365-3083.2011.02597.x. PubMed

Janda A, Rensing-Ehl A, Eibel H, Ehl S, Speckmann C.

29. OKTOBER 2011 (PID-NET)

Concise review: managing genotoxicity in the therapeutic modification of stem cells.

Stem Cells. 2011 Oct;29(10):1479-84. doi: 10.1002/stem.716. Review. PubMed

Baum C, Modlich U, Göhring G, Schlegelberger B.

19. OKTOBER 2011 (PID-NET)

Correction of murine SCID-X1 by lentiviral gene therapy using a codon-optimized IL2RG gene and minimal pretransplant conditioning.

Mol Ther. 2011 Oct;19(10):1867-77. doi: 10.1038/mt.2011.127. Epub 2011 Jul 12. PubMed

Huston MW, van Til NP, Visser TP, Arshad S, Brugman MH, Cattoglio C, Nowrouzi A, Li Y, Schambach A, Schmidt M, Baum C, von Kalle C, Mavilio F, Zhang F, Blundell MP, Thrasher AJ, Verstegen MM, Wagemaker G.

1. OKTOBER 2011 (PID-NET)

Clinical and immunological manifestations of patients with atypical severe combined immunodeficiency.

Clin Immunol.,141, 73-82, 2011.

Felgentreff, K., Perez-Becker, R., Speckmann, C., Schwarz, K., Kalwak, K., Markelj, G., Avcin, T., Qasim, W., Davies, E.G., Niehues, T. und Ehl, S.

25. SEPTEMBER 2011 (PID-NET)

Correction of murine Rag1 deficiency by self-inactivating lentiviral vector-mediated gene transfer.

Leukemia. 2011 Sep;25(9):1471-83. doi: 10.1038/leu.2011.106. Epub 2011 May 27. PubMed

Pike-Overzet K, Rodijk M, Ng YY, Baert MR, Lagresle-Peyrou C, Schambach A, Zhang F, Hoeben RC, Hacein-Bey-Abina S, Lankester AC, Bredius RG, Driessen GJ, Thrasher AJ, Baum C, Cavazzana-Calvo M, van Dongen JJ, Staal FJ.

1. SEPTEMBER 2011 (PID-NET)

Redundant ability of phagocytes to kill Aspergillus species.

J Allergy Clin Immunol 128(3):686-7.

Roesler J und Rösen-Wolff A

31. AUGUST 2011 (PID-NET)

Development of novel efficient SIN vectors with improved safety features for Wiskott-Aldrich syndrome stem cell based gene therapy.

Mol Pharm. 2011 Oct 3;8(5):1525-37. Epub 2011 Aug 31. PubMed

Avedillo Díez I, Zychlinski D, Coci EG, Galla M, Modlich U, Dewey RA, Schwarzer A, Maetzig T, Mpofu N, Jaeckel E, Boztug K, Baum C, Klein C, Schambach A.

1. AUGUST 2011 (PID-NET)

Development of novel efficient SIN vectors with improved safety features for Wiskott-Aldrich syndrome stem cell based gene therapy.

Molecular pharmaceutics. 2011;8(5):1525-37.

Avedillo Diez I, Zychlinski D, Coci EG, Galla M, Modlich U, Dewey RA, Schwarzer A, Maetzig T, Mpofu N, Jaeckel E, Boztug K, Baum C, Klein C, Schambach A.

8. JULI 2011 (PID-NET)

Diagnostik primärer Immundefekte. Vom klinisch-immunologischen Phänotyp zur molekularen Diagnose.

Monatsschrift für Kinderheilkunde, 159, 434-442, 2011.

Von Bernuth, H., Schwarz, K., Renner, E.D. und Ehl, S.

1. JULI 2011 (PID-NET)

G6PC3 mutations are associated with a major defect of glycosylation: a novel mechanism for neutrophil dysfunction.

Glycobiology. 2011;21(7):914-24.

Hayee B, Antonopoulos A, Murphy EJ, Rahman FZ, Sewell G, Smith BN, McCartney S, Furman M, Hall G, Bloom SL, Haslam SM, Morris HR, Boztug K, Klein C, Winchester B, Pick E, Linch DC, Gale RE, Smith AM, Dell A, Segal AW.

1. APRIL 2011 (PID-NET)

Curative treatment of two patients with combined immunodeficiency syndrome and DOCK8 mutations by hematopoietic cell transplantation.

Bone Marrow Transplantation, 46, 552-556, 2011

Gatz, S.A., Benninghoff, U., Schütz, C., Schulz, A., Hönig, M., Pannicke, U., Holzmann, K.-H., Schwarz, K. und Friedrich, W.

22. MÄRZ 2011 (PID-NET)

Promoter competition for gene therapy of SCID-X1.

Hum Gene Ther. 2011 Mar;22(3):255-6. PubMed

Baum C, Schambach A.

1. MÄRZ 2011 (PID-NET)

A case of syndromic neutropenia and mutation in G6PC3.

Journal of pediatric hematology/oncology. 2011;33(2):138-40.

Gatti S, Boztug K, Pedini A, Pasqualini C, Albano V, Klein C, Pierani P.

1. MÄRZ 2011 (PID-NET)

Meningoencephalitis caused by varicella zoster virus reactivation in a child with dominant partial interferon-gamma receptor-1 deficiency.

Pediatr Infect Dis J 30 (3): 265-266.

Roesler J, Hedrich C, Laass MW, Heyne K, Rösen-Wolff A

1. MÄRZ 2011 (PID-NET)

Hyperbilirubinemia, cholestatic hepatitis and fetal hepatic failure in TlowB-NK- Severe Combined Immunodeficiency caused by Adenosine Deaminase Deficiency (ADA-SCID).

Klinische Pädiatrie, 223, 85-89, 2011.

Kühl, J.-S., Schwarz, K., Münche, A., Schmutz, M., Pekrun, A., Meisel, C., Wahn, V., Ebell, W. und von Bernuth, H.

23. FEBRUAR 2011 (PID-NET)

Genetic etiologies of severe congenital neutropenia.

Current Opinion in Pediatrics. 2011;23(1):21-6.

Boztug K, Klein C

19. FEBRUAR 2011 (PID-NET)

Solving the problem of γ -retroviral vectors containing long terminal repeats.

Mol Ther. 2011 Feb;19(2):229-31. PubMed

Persons DA, Baum C.

3. FEBRUAR 2011 (PID-NET)

Parachuting in the epigenome: the biology of gene vector insertion profiles in the context of clinical trials.

EMBO Mol Med. 2011 Feb;3(2):75-7. doi: 10.1002/emmm.201000110. Epub 2011 Jan 21. PubMed

Baum C.

20. JANUAR 2011 (PID-NET)

PD-L1 blockade effectively restores strong graft-versus-leukemia effects without graft-versus-host disease after delayed adoptive transfer of T-cell receptor gene-engineered allogeneic CD8(+) T cells.

Blood. 2011;117(3):1030-41.

Koestner W, Hapke M, Herbst J, Klein C, Welte K, Fruehauf J, Flatley A, Vignali DA, Hardtke-Wolenski M, Jaeckel E, Blazar BR, Sauer MG.

5. JANUAR 2011 (PID-NET)

Defective IL10 Signaling Defining a Subgroup of Patients With Inflammatory Bowel Disease.

American Journal of Gastroenterology. 2011;106(8):1544-55.

Begue B, Verdier J, Rieux-Laucat F, Goulet O, Morali A, Canioni D, Hugot J-P, Daussy C, Verkarre V, Pigneur B, Fischer A, Klein C, Cerf-Bensussan N, Ruemmele FM.