

Thematic Network: Research for Rare Diseases and Personalised Medicine (2001-2021)

So far published papers with our project partners:

Christoph Klein (LMU), Scott Snapper (Harvard Medical School), Alexio Muise (University of Toronto), Raz Somech (Sheba Academic Medical Center Hospital, Tel Aviv), Ekrem Ünal (Eriyces University, Kayseri), Roya Sherkat (Isfahan University) and Josef Penninger (IMBA, Wien):

A crucial role for Jagunal homolog 1 in humoral immunity and antibody glycosylation in mice and humans.

J Exp Med. 2021 Jan 4;218(1):e20200559. [doi: 10.1084/jem.20200559](https://doi.org/10.1084/jem.20200559).

Hagelkruys A, Wirnsberger G, Stadlmann J, Wöhner M, Horrer M, Vilagos B, Jonsson G, Kogler M, Tortola L, Novatchkova M, Bönelt P, Hoffmann D, Koglgruber R, Steffen U, Schett G, Busslinger M, Bergthaler A, Klein C, Penninger JM.

Clinical Genomics for the Diagnosis of Monogenic forms of Inflammatory Bowel Disease: A Position Paper from The Paediatric IBD Porto Group of ESPGHAN.

J Pediatr Gastroenterol Nutr. 2020 Dec 16; Publish Ahead of Print. [doi:](https://doi.org/10.1097/MPG.0000000000003017)

[10.1097/MPG.0000000000003017](https://doi.org/10.1097/MPG.0000000000003017). [Epub ahead of print].

Uhlig HH, Charbit-Henrion F, Kotlarz D, Shouval DS, Schwerd T, Strisciuglio C, de Ridder L, van Limbergen J, Macchi M, Snapper SB, Ruemmele FM, Wilson DC, Travis SPL, Griffiths AM, Turner D, Klein C, Muise AM, Russell RK; Paediatric IBD Porto group of ESPGHAN.

The E3 ubiquitin ligase UBR5 interacts with TTC7A and may be associated with very early onset inflammatory bowel disease.

Sci Rep. 2020 Oct 29;10(1):18648. [doi: 10.1038/s41598-020-73482-6](https://doi.org/10.1038/s41598-020-73482-6).

Dhingani N, Guo C, Pan J, Li Q, Warner N, Jardine S, Leung G, Kotlarz D, Gonzaga-Jauregui C, Klein C, Snapper SB, Navas-López VM, Muise AM.

Common Variable Immunodeficiency, Autoimmune Hemolytic Anemia, and Pancytopenia Associated With a Defect in IKAROS.

J Pediatr Hematol Oncol. 2020 Oct 28. [doi: 10.1097/MPH.0000000000001976](https://doi.org/10.1097/MPH.0000000000001976).

Yilmaz E, Kuehn HS, Odakir E, Niemela JE, Ozcan A, Eken A, Rohlf M, Cansever M, Gok V, Aydin F, Karakukcu M, Hauck F, Klein C, Unal E, Rosenzweig SD, Papiroglu T.

An RTEL1 Mutation Links to Infantile-Onset Ulcerative Colitis and Severe Immunodeficiency.

J Clin Immunol. 2020 Oct;40(7):1010-1019. [doi: 10.1007/s10875-020-00829-z](https://doi.org/10.1007/s10875-020-00829-z). Epub 2020 Jul 24.

Ziv A, Werner L, Konnikova L, Awad A, Jeske T, Hastreiter M, Mitsialis V, Stauber T, Wall S, Kotlarz D, Klein C, Snapper SB, Tzfati Y, Weiss B, Somech R, Shouval DS.

Type 1 Plasminogen Deficiency With Pulmonary Involvement: Novel Treatment and Novel Mutation.

J Pediatr Hematol Oncol. 2020 Sep 15. [doi: 10.1097/MPH.0000000000001951](https://doi.org/10.1097/MPH.0000000000001951).

Hangul M, Tuzuner AB, Somekh I, Klein C, Papiroglu T, Unal E, Kose M.

Predictive Prenatal Diagnosis for Infantile-Onset Inflammatory Bowel Disease due to Interleukin-10 Signalling Defects.

Pediatr Gastroenterol Nutr. 2020 Sep 9. [doi: 10.1097/MPG.0000000000002937](https://doi.org/10.1097/MPG.0000000000002937). [Epub ahead of print].

Ye Z, Hu W, Wu B, Zhang Y, Lei C, Williams I, Shouval D, Kanegane H, Kim KM, de Ridder L, Shah N, Ling G, Yerushalmi B, Kotlarz D, Snapper S, Horn R, Klein C, Muise A, Huang Y, Uhlig HH.

Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases.

Gene. 2020 Jul 30;749:144709. [doi: 10.1016/j.gene.2020.144709](https://doi.org/10.1016/j.gene.2020.144709).

Boonsimma P, Michael Gasser M, Netbaramee W, Wechapinan T, Srichomthong C, Ittiwut C, Wagner M, Krenn M, Zimprich F, Abicht A, Biskup S, Roser T, Borggraefe I, Suphapeetiporn K, Shotelersuk V.

An RTEL1 Mutation Links to Infantile-Onset Ulcerative Colitis and Severe Immunodeficiency

Journal of Clinical Immunology. 2020 Jul 24. doi: [10.1007/s10875-020-00829-z](https://doi.org/10.1007/s10875-020-00829-z). Online ahead of print. *J Clin Immunol*. 2020.

Ziv A, Werner L, Konnikova L, Awad A, Jeske T, Hastreiter M, Mitsialis V, Stauber T, Wall S, Kotlarz D, Klein C, Snapper SB, Tzfati Y, Weiss B, Somech R, Shouval DS .

NOX1 Regulates Collective and Planktonic Cell Migration: Insights From Patients With Pediatric-Onset IBD and NOX1 Deficiency.

Inflamm Bowel Dis. 2020 Jul 17;26(8):1166-1176. doi: [10.1093/ibd/izaa017](https://doi.org/10.1093/ibd/izaa017).

Khoshnevisan R, Anderson M, Babcock S, Anderson S, Illig D, Marquardt B, Sherkat R, Schröder K, Moll F, Hollizeck S, Rohlf M, Walz C, Adibi P, Rezaei A, Andalib A, Koletzko S, Muise AM, Snapper SB, Klein C, Thiagarajah JR, Kotlarz D.

Prevalence and Clinical Features of Inflammatory Bowel Diseases Associated With Monogenic Variants, Identified by Whole-Exome Sequencing in 1000 Children at a Single Center

Gastroenterology. 2020 Jun; 158(8):2208-2220. doi: [10.1053/j.gastro.2020.02.023](https://doi.org/10.1053/j.gastro.2020.02.023). Epub 2020 Feb 19.

Crowley E, Warner N, Pan J, Khalouei S, Elkadri A, Fiedler K, Foong J, Turinsky AL, Bronte-Tinkew D, Zhang S, Hu J, Tian D, Li D, Horowitz J, Siddiqui I, Upton J, Roifman CM, Church PC, Wall DA, Ramani AK, Kotlarz D, Klein C, Uhlig H, Snapper SB, Gonzaga-Jauregui C, Paterson AD, McGovern DPB, Brudno M, Walters TD, Griffiths AM, Muise AM.

Very Early Onset Inflammatory Bowel Disease: A Clinical Approach With a Focus on the Role of Genetics and Underlying Immune Deficiencies.

Inflamm Bowel Dis. 2020 May 12;26(6):820-842. doi: [10.1093/ibd/izz259](https://doi.org/10.1093/ibd/izz259).

Ouahed J, Spencer E, Kotlarz D, Shouval DS, Kowalik M, Peng K, Field M, Grushkin-Lerner L, Pai SY, Bousvaros A, Cho J, Argmann C, Schadt E, McGovern DPB, Mokry M, Nieuwenhuis E, Clevers H, Powrie F, Uhlig H, Klein C, Muise A, Dubinsky M, Snapper SB.

Whole exome sequencing (WES) Approach for diagnosing Primary immunodeficiencies (PIDs) in a highly consanguineous community.

Clinical Immunology. 2020 May;214:108376. [doi: 10.1016/j.clim.2020.108376](https://doi.org/10.1016/j.clim.2020.108376). Epub 2020 Mar 3.

Simon AJ, Golan AC, Lev A, Stauber T, Barel O, Somekh I, Klein C, AbuZaitun O, Eyal E, Kol N, Unal E, Amariglio N, Rechavi G, Somech R.

Author Correction: Human FCHO1 deficiency reveals role for clathrin-mediated endocytosis in development and function of T cells.

Nature Communications. 2020 Apr 20;11(1):1963. [doi: 10.1038/s41467-020-15946-x](https://doi.org/10.1038/s41467-020-15946-x).

Lyszkiewicz M, Ziętara N, Frey L, Pannicke U, Stern M, Liu Y, Fan Y, Puchalka J, Hollizeck S, Somekh I, Rohlf s M, Yilmaz T, Ünal E, Karakukcu M, Patiroğlu T, Kellerer C, Karasu E, Sykora KW, Lev A, Simon A, Somech R, Roesler J, Hoenig M, Keppler OT, Schwarz K, Klein C.

A Rare Case of Activated Phosphoinositide 3-Kinase Delta Syndrome (APDS) Presenting With Hemophagocytosis Complicated With Hodgkin Lymphoma.

J Pediatr Hematol Oncol. 2020 Mar;42(2):156-159. [doi: 10.1097/MPH.0000000000001487](https://doi.org/10.1097/MPH.0000000000001487).

Cansever M, Zietara N, Chiang SCC, Ozcan A, Yilmaz E, Karakukcu M, Rohlf s M, Somekh I, Canoz O, Abdulrezzak U, Bryceson Y, Klein C, Unal E, Patiroglu T.

Refractory and Fatal Presentation of Severe Autoimmune Hemolytic Anemia in a Child With the DNASE1L3 Mutation Complicated With an Additional DOCK8 Variant.

Journal of Pediatric Hematology and Oncology. 2020 Mar 20. [doi:](https://doi.org/10.1097/MPH.0000000000001780)

[10.1097/MPH.0000000000001780](https://doi.org/10.1097/MPH.0000000000001780). [Epub ahead of print].

Paç Kisaarslan A, Witzel M, Unal E, Rohlf s M, Akyildiz B, Dogan ME, Poyrazoglu H, Klein C, Patiroglu T.

Drug Screen Identifies Leflunomide for Treatment of Inflammatory Bowel Disease Caused by TTC7A Deficiency.

Gastroenterology. 2020 Mar;158:1000-1015. [doi: 10.1053/j.gastro.2019.11.019](https://doi.org/10.1053/j.gastro.2019.11.019).

Jardine S, Anderson S, Babcock S, Leung G, Pan J, Dhingani N, Warner N, Guo C, Siddiqui I, Kotlarz D, Dowling JJ, Melnyk RA, Snapper SB, Klein C, Thiagarajah JR, Muise AM.

ATP1A3-related epilepsy: Report of seven cases and literature-based analysis of treatment response.

J Clin Neurosci. 2020 Feb;72:31-38. [doi: 10.1016/j.jocn.2020.01.041](https://doi.org/10.1016/j.jocn.2020.01.041).

Gasser M, Boonsimma P, Netbaramee W, Wechapinan T, Srichomthong C, Ittiwut C, Krenn M, Zimprich F, Milenkovic I, Abicht A, Biskup S, Roser T, Shotelersuk V, Tacke M, Kuersten M, Wagner M, Borggraefe I, Suphapeetiporn K, von Stülpnagel C.

NOX1 Regulates Collective and Planktonic Cell Migration: Insights From Patients With Pediatric-Onset IBD and NOX1 Deficiency.

Inflammatory Bowel Disease. 2020 Feb 17. pii: [izaa017](https://doi.org/10.1093/ibd/izaa017). [doi: 10.1093/ibd/izaa017](https://doi.org/10.1093/ibd/izaa017). [Epub ahead of print].

Khoshnevisan R, Anderson M, Babcock S, Anderson S, Illig D, Marquardt B, Sherkat R, Schröder K, Moll F, Hollizeck S, Rohlf s M, Walz C, Adibi P, Rezaei A, Andalib A, Koletzko S, Muise AM, Snapper SB, Klein C, Thiagarajah JR, Kotlarz D.

Human FCHO1 deficiency reveals role for clathrin-mediated endocytosis in development and function of T cells.

Nature Communications. 2020 Feb 25;11(1):1031. [doi: 10.1038/s41467-020-14809-9](https://doi.org/10.1038/s41467-020-14809-9).

Lyszkiewicz M, Zię t ara N, Frey L, Pannicke U, Stern M, Liu Y, Fan Y, Puchałka J, Hollizeck S, Somekh I, Rohlf s M, Yilmaz T, Ünal E, Karakukcu M, Patirođlu T, Kellerer C, Karasu E, Sykora KW, Lev A, Simon A, Somech R, Roesler J, Hoenig M, Keppler OT, Schwarz K, Klein C.

A case report of sinusoidal diffuse large B-cell lymphoma in a STK4 deficient Patient.

Medicine (Baltimore). 2020 Feb;99(9):e18601. [doi: 10.1097/MD.00000000000018601](https://doi.org/10.1097/MD.00000000000018601).

Ashrafi F, Klein C, Poorpooneh M, Sherkat R, Khoshnevisan R.

Correction to: Human Inborn Errors of Immunity: 2019 Update on the Classification from the International Union of Immunological Societies Expert Committee.

J Clin Immunol. 2020 Jan;40(1):65. [doi: 10.1007/s10875-020-00763-0](https://doi.org/10.1007/s10875-020-00763-0).

Tangye SG, Al-Herz W, Bousfiha A, Chatila T, Cunningham-Rundles C, Etzioni A, Franco JL, Holland SM, Klein C, Morio T, Ochs HD, Oksenhendler E, Picard C, Puck J, Torgerson TR, Casanova JL, Sullivan KE.

Human Inborn Errors of Immunity: 2019 Update of the IUIS Phenotypical Classification.

J Clin Immunol. 2020 Jan;40(1):66-81. doi: [10.1007/s10875-020-00758-x](https://doi.org/10.1007/s10875-020-00758-x). Epub 2020 Feb 11.
Bousfiha A, Jeddane L, Picard C, Al-Herz W, Ailal F, Chatila T, Cunningham-Rundles C, Etzioni A, Franco JL, Holland SM, Klein C, Morio T, Ochs HD, Oksenhendler E, Puck J, Torgerson TR, Casanova JL, Sullivan KE, Tangye SG.

Very Early Onset Inflammatory Bowel Disease: A Clinical Approach With a Focus on the Role of Genetics and Underlying Immune Deficiencies.

Inflammatory Bowel Disease. 2019, pii: izz259, doi: [10.1093/ibd/izz259](https://doi.org/10.1093/ibd/izz259). [Epub ahead of print]

Ouahed J, Spencer E, Kotlarz D, Shouval DS, Kowalik M, Peng K, Field M, Grushkin-Lerner L, Pai SY, Bousvaros A, Cho J, Argmann C, Schadt E, McGovern DPB, Mokry M, Nieuwenhuis E, Clevers H, Powrie F, Uhlig H, Klein C, Muise A, Dubinsky M, Snapper SB.

Drug Screen Identifies Leflunomide for Treatment of Inflammatory Bowel Diseases Caused by TTC7A Deficiency.

Gastroenterology. 2019, 19, 41574-6, doi: [10.1053/j.gastro.2019.11.019](https://doi.org/10.1053/j.gastro.2019.11.019). [Epub ahead of print]
Jardine S, Anderson S, Babcock S, Leung G, Pan J, Dhingani N, Warner N, Guo C, Siddiqui I, Kotlarz D, Dowling JJ, Melnyk R, Snapper SB, Klein C, Thiagarajah JR, Muise AM.

CD137 deficiency causes immune dysregulation with predisposition to lymphomagenesis.

Blood. 2019, 134(18),1510-1516, doi: [10.1182/blood.2019000644](https://doi.org/10.1182/blood.2019000644).
Somekh I, Thian M, Medgyesi D, Gülez N, Magg T, Gallón Duque A, Stauber T, Lev A, Genel F, Unal E, Simon AJ, Lee YN, Kalinichenko A, Dmytrus J, Kraakman MJ, Schiby G, Rohlfms M, Jacobson JM, Özer E, Akcal Ö, Conca R, Patiroglu T, Karakukcu M, Ozcan A, Shahin T, Appella E, Tatematsu M, Martinez-Jaramillo C, Chinn IK, Orange JS, Trujillo-Vargas CM, Franco JL, Hauck F, Somech R, Klein C, Boztug K.

IL-12R β 1 deficiency corresponding to concurrency of two diseases, medelian susceptibility to mycobacterial disease and Crohen's disease.

Journal of clinical tuberculosis and other mycobacterial diseases. 2019, 17:100123, [doi: 10.1016/j.jctube.2019.100123](https://doi.org/10.1016/j.jctube.2019.100123), eCollection 2019 Dec.

Khoshnevisan R, Nekooei-Marnany N, Klein C, Kotlarz D, Behnam M, Ostadi V, Yaran M, Rezaei A, Sherkat R.

Fetuin-A deficiency is associated with infantile cortical hyperostosis (Caffey disease).

Pediatric Research. 2019, [doi: 10.1038/s41390-019-0499-0](https://doi.org/10.1038/s41390-019-0499-0). [Epub ahead of print]

Merdler-Rabinowicz R, Grinberg A, Jacobson JM, Somekh I, Klein C, Lev A, Ihsan S, Habib A, Somech R, Simon AJ.

Genetic Deficiency and Biochemical Inhibition of ITK Affect Human Th17, Treg, and Innate Lymphoid Cells.

Journal of Clinical Immunology. 2019, 39, 391. doi.org/10.1007/s10875-019-00632-5.

Eken A, Cansever M, Somekh I, Mizoguchi Y, Zietara N, Okus FZ, Erdem S, Canatan H, Akyol S, Ozcan A, Karakukcu M, Hollizeck S, Rohlfs M, Unal E, Klein C, Patiroglu T.

CARMIL2 Deficiency Presenting as Very Early Onset Inflammatory Bowel Disease.

Inflammatory Bowel Disease. 2019, pii: izz103, [doi: 10.1093/ibd/izz103](https://doi.org/10.1093/ibd/izz103).

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, Rohls M, Koletzko S, Lurz E, Muise AM, Snapper SB, Hauck F, Klein C, Kotlarz D.

A Rare Case of Activated Phosphoinositide 3-Kinase Delta Syndrome (APDS) Presenting With Hemophagocytosis Complicated With Hodgkin Lymphoma

Journal of Pediatric Hematology/Oncology. 2019, [doi: 10.1097/MPH.0000000000001487](https://doi.org/10.1097/MPH.0000000000001487).

Cansever M, Zietara N, Chiang SCC, Ozcan A, Yilmaz E, Karakukcu M, Rohlfs M, Somekh I, Canoz O, Abdulrezzak U, Bryceson Y, Klein C, Unal E, Patiroglu T.

Human RIPK1 deficiency causes combined immunodeficiency and inflammatory bowel diseases

Proceedings of the National Academy of Sciences. 2019, 116 (3), 970-975, [doi: 10.1073/pnas.1813582116](https://doi.org/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 s M, Conca R, Field M, Warmer N, Mordechai S, Shteyer E, Turner D, Boukari R, Belbouab R, Walz C, Gaid MM, Hornung V, Baumann B, Pannicke U, Al Idrissi E, Ali Alghamadi H, Sepulveda FE, Gil M, de Saint Basile G, Hönig M, Koletzko S, Muise AM, Snapper SB, Schwarz K, Klein C, Kotlarz D.

Proteome Analysis of human neutrophil granulocytes from patients with monogenic disease using data-independent acquisition

Molecular & Cellular Proteomics. 2019, 4, 760-772. [doi: 10.1074/mcp.RA118.001141](https://doi.org/10.1074/mcp.RA118.001141).

Grabowski P, Hesse S, Hollizeck S, Rohlf s M, Behrends U, Sherkat R, Tamary H, Ünal E, Somech R, Patiroğlu T, Canzar S, van der Werff Ten Bosch J, Klein C, Rappsilber J.

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

Gastroenterology. 2018, 18, 35036-4, pii: S0016-5085(18)35036-4, [doi: 10.1053/j.gastro.2018.09.041](https://doi.org/10.1053/j.gastro.2018.09.041).

Lehle AS, Farin HF, Marquardt B, Michels BE, Magg T, Li Y, Liu Y, Ghalandary M, Lam mens K, Hollizeck S, Rohlf s M, Hauck F, Conca R, Walz C, Weiss B, Lev A, Simon AJ, Groß O, Gaidt MM, Hornung V, Clevers H, Yazbeck N, Hanna-Wakim R, Shouval DS, Warner N, Somech R, Muise AM, Snapper SS, Bufler P, Koletzko S, Klein C, Kotlarz D.

Novel Mutations in *RASGRP1* are Associated with Immunodeficiency, Immune Dysregulation, and EBV-Induced Lymphoma

Journal of Clinical Immunology. 2018, 38(6), 699-710, [doi: 10.1007/s10875-018-0533-8](https://doi.org/10.1007/s10875-018-0533-8). Epub 2018 Jul 20.

Somekh I, Marquardt B, Liu Y, Rohlf s M, Hollizeck S, Karakukcu M, Unal E, Yilmaz E, Patiroğlu T, Cansever M, Frizinsky S, Vishnvenska-Dai V, Rechavi E, Stauber T, Simon AJ, Lev A, Klein C, Kotlarz D, Somech R.

Human TGF- β 1 deficiency causes severe inflammatory bowel disease and encephalopathy.

Nature Genetics. 2018, 50(3), 344-348, [doi: 10.1038/s41588-018-0063-6](https://doi.org/10.1038/s41588-018-0063-6).

Kotlarz D, Marquardt B, Barøy T, Lee WS, Konnikova L, Hollizeck S, Magg T, Lehle AS, Walz C, Borggraefe I, Hauck F, Bufler P, Conca R, Wall SM, Schumacher EM, Misceo D, Frengen E, Bentsen BS, Uhlig HH, Hopfner KP, Muise AM, Snapper SB, Strømme P, Klein C.

Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity.

Journal of Clinical Immunology. 2018, 38(1), 96-128, [doi: 10.1007/s10875-015-0201-1](https://doi.org/10.1007/s10875-015-0201-1).

Picard C, Bobby Gaspar H, Al-Herz W, Bousfiha A, Casanova JL, Chatila T, Crow YJ, Cunningham-Rundles C, Etzioni A, Franco JL, Holland SM, Klein C, Morio T, Ochs HD, Oksenhendler E, Puck J, Tang MLK, Tangye SG, Torgerson TR, Sullivan KE.

The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies.

Journal of Clinical Immunology. 2018, 38(1), 129-143, [doi: 10.1007/s10875-017-0465-8](https://doi.org/10.1007/s10875-017-0465-8). Epub 2017 Dec 11.

Bousfiha A, Jeddane L, Picard C, Ailal F, Bobby Gaspar H, Al-Herz W, Chatila T, Crow YJ, Cunningham-Rundles C, Etzioni A, Franco JL, Holland SM, Klein C, Morio T, Ochs HD, Oksenhendler E, Puck J, Tang MLK, Tangye SG, Torgerson TR, Casanova JL, Sullivan KE.

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

Inflammatory Bowel Disease. 2017, 23(11), 1950-1961, [doi: 10.1097/MIB.0000000000001270](https://doi.org/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.

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

Journal of Clinical Immunology. 2017, 140(4), 1112-1119, [doi: 10.1016/j.jaci.2016.10.053](https://doi.org/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.

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

Nature Genetics. 2017, 49, 742-752, [doi: 10.1038/ng.3833](https://doi.org/10.1038/ng.3833).

Witzel M, Petersheim D, Fan Y, Bahrami E, Racek T, Rohlf M, Puchałka 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.

EBV lymphoproliferative-associated disease and primary cardiac T-cell lymphoma in a STK4 deficient patient: A case report.

Medicine (Baltimore). 2017, 96(48), e8852, [doi: 10.1097/MD.00000000000008852](https://doi.org/10.1097/MD.00000000000008852).

Sherkat R, Sabri MR, Dehghan B, Bigdelian H, Reisi N, Afsharmoghadam N, Rahimi H, Rahmanian N, Klein C.

“Inherited IL-17RA deficiency in patients with fungal and bacterial diseases: genetic, immunological, and clinical Features”

Proceedings of the National Academy of Sciences of the United States of America (PNAS).

2016, [doi: 10.1073/pnas.1618300114](https://doi.org/10.1073/pnas.1618300114).

Lévy R, Okada S, Beziat V, Moriya K, Liu C, Chai LYA, Migaud M, Gothe F, Al Ali A, Cyrus C, Vatte C, Patisroglu T, Unal E, Ferneiny M, Hyakuna N, Nepesov S, Oleastro M, Ikin-ciogullari A, Dogu F, Asano T, Ohara O, Yun L, Della Mina E, Bronnimann D, Itan Y, Klein C, Bustamante J, Boisson-Dupuis S, Tahuil N, Aytakin C, Salhi A, Al Muhsen S, Kobayashi M, Toubiana J, Abel L, Li X, Camcioglu Y, Celmeli F, Hauck F, Al-Khater SA, Casanova JL, Puel A.

Interleukin 1 β Mediates Intestinal Inflammation in Mice and Patients With Interleukin 10 Receptor Deficiency.

Gastroenterology. 2016, 151(6), 1100-1104, doi: 10.1053/j.gastro.2016.08.055. Epub 2016 Sep 28.

Shouval DS, Biswas A, Kang YH, Griffith AE, Konnikova L, Mascanfroni ID, Redhu NS, Frei SM, Field M, Doty AL, Goldsmith JD, Bhan AK, Loizides A, Weiss B, Yerushalmi B, Yanagi T, Lui X, Quintana FJ, Muise AM, Klein C, Horwitz BH, Glover SC, Bousvaros A, Snapper SB.

Interleukin 1 β Mediates Intestinal Inflammation in Mice and Patients With Interleukin 10 Receptor Deficiency.

Gastroenterology. 2016, 151(6), 1100-1104, doi: 10.1053/j.gastro.2016.08.055. Epub 2016 Sep 28.

Shouval DS, Biswas A, Kang YH, Griffith AE, Konnikova L, Mascanfroni ID, Redhu NS, Frei SM, Field M, Doty AL, Goldsmith JD, Bhan AK, Loizides A, Weiss B, Yerushalmi B, Yanagi T, Lui X, Quintana FJ, Muise AM, Klein C, Horwitz BH, Glover SC, Bousvaros A, Snapper SB.

Large B-Cell Lymphoma in an Adolescent Patient with IL-10 Receptor Deficiency and History of Infantile Inflammatory Bowel Disease.

Journal of Pediatric Gastroenterology and Nutrition. 2016, 63(1), e15–e17.

Shouval DS, Ebens CL, Murchie R, McCann K, Rabah R, Klein C, Muise A, Snapper SB.

FOXP3⁺ Tregs require WASP to restrain Th2-mediated food allergy.

Journal of Clinical Investigation. 2016, 126(10), 4030-4044, doi: 10.1172/JCI85129. Epub 2016 Sep 19

Lexmond WS, Goettel JA, Lyons JJ, Jacobse J, Deken MM, Lawrence MG, DiMaggio TH, Kotlarz D, Garabedian E, Sackstein P, Nelson CC, Jones N, Stone KD, Candotti F, Rings EH, Thrasher AJ, Milner JD, Snapper SB, Fiebiger E.

Variants in TRIM22 That Affect NOD2 Signaling Are Associated With Very-Early-Onset Inflammatory Bowel Disease.

Gastroenterology. 2016, 150(5), 1196-207, doi: 10.1053/j.gastro.2016.01.031. Epub 2016 Feb 4. PMID: 26836588

Li Q, Lee CH, Peters LA, Mastropaolo LA, Thoeni C, Elkadri A, Schwerd T, Zhu J, Zhang B, Zhao Y, Hao K, Dinarzo A, Hoffman G, Kidd BA, Murchie R, Al Adham Z, Guo C, Kotlarz D, Cutz E, Walters TD, Shouval DS, Curran M, Dobrin R, Brodmerkel C, Snapper SB, Klein C, Brumell JH, Hu M, Nanan R, Snanter-Nanan B, Wong M, Le Deist F, Haddad E, Roifman CM, Deslandres C, Griffiths AM, Gaskin KJ, Uhlig HH, Schadt EE, Muise AM.

Clinical features and genetic analysis of six patients with Wiskott-Aldrich Syndrome reporting two novel mutations: Experience of Erciyes University, Kayseri, Turkey.

Journal of Genetic Counseling, 2016, 27(1), 9-24.

Patiroglu T, Klein C, Gungor HE, Ozdemir MA, Witzel M, Karakukcu M, Sawalle-Belohradsky J, Conca R, Unal E.

Severe congenital neutropenia with neurological impairment due to a homozygous VPS45 p.E238K mutation: A case report suggesting a genotype-phenotype correlation.

American Journal of Medical Genetics Part A. 2015, 167A(12), 3214-8,

doi: 10.1002/ajmg.a.37367. Epub 2015 Sep 11. PMID: 26358756

Meerschaut I, Bordon V, Dhooge C, Delbeke P, Vanlander AV, Simon A, Klein C, Kooy RF, Somech R, Callewaert B.

Fatal autoimmunity in mice reconstituted with human hematopoietic stem cells encoding defective foxp3.

Blood. 2015, 125, 3886-3895.

Goettel JA, Biswas S, Lexmond WS, Yeste A, Passerini L, Patel B, Yang S, Sun J, Ouahed J, Shouval Ds, McCann KJ, Horwitz BH, Mathis D, Milford EL, Notarangelo LD, Roncarolo MG, Fiebiger E, Marasco WA, Bacchetta R, Quintana FJ, Pai SY, Klein C, Muise AM, Snapper SB.

Very early-onset inflammatory bowel disease: Gaining insight through focused discovery.

Inflammatory Bowel Disease. 2015, 21, 1166-1175.
Moran CJ, Klein C, Muise AM, Snapper SB.

Mutations in Tetratricopeptide Repeat Domain 7A Result in a Severe Form of Very Early Onset Inflammatory Bowel Disease.

Gastroenterology. 2014, 146, 1028-1033.
Avitzur Y, Guo C, Mastropaolo LA, Bahrami E, Chen H, Zhao Z, Elkadri A, Dhillon S, Murchie R, Fattouh R, Huynh H, Walker JL, Wales PW, Cutz E, Kakuta Y, Dudley J, Kammermeier J, Powrie F, Shah N, Walz C, Nathrath M, Kotlarz D, Puchaka J, Krieger J, Racek T, Kirchner T, Walters TD, Brumell JH, Griffiths AM, Rezaei N, Rashtian P, Najafi M, Monajemzadeh M, Pelsue S, McGovern DP, Uhlig HH, Schadt E, Klein C, Snapper SB, Muise AM.

Very Early Onset Inflammatory Bowel Disease Associated with Aberrant Trafficking of IL-10R1 and Cure by T Cell Replete Haploidentical Bone Marrow Transplantation.

Journal of Clinical Immunology. 2014, 34,331-9.
Murugan D, Albert MH, Langemeier J, Bohne J, Puchalka J, Jarvinen PM, Hauck F, Klenk AK, Prell C, Schatz S, Diestelhorst J, Sciskala B, Kohistani N, Belohradsky BH, Muller S, Kirchner T, Walter MR, Bufler P, Muise AM, Snapper SB, Koletzko S, Klein C, Kotlarz D.

IL-10 Receptor Signaling in Innate Immune Cells Regulates Mucosal Immune Tolerance and Anti-Inflammatory Macrophage Function Immunity.

Immunity. 2014, 40(5), 706-19.
Shouval DS, Biswas A, Goettel JA, McCann K, Ibourk M, Conaway E, Lavoie S, Nguyen DD, Samsom JN, Somech R, Weiss B, Beier R, Conklin L, Bhan AK, Mora JR, Klein C, Muise AM, Horwitz BH, Snapper SB.

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

Journal of Allergy and Clinical Immunology. 2014, 131(3), 825-30.

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.

Interleukin 10 receptor signaling: master regulator of intestinal mucosal homeostasis in mice and humans.

Advanced Immunology. 2014, 122, 177-210.

Shouval DS, Ouahed J, Biswas A, Goettel JA, Horwitz BH, Klein C, Muise AM, Snapper SB.

The Diagnostic Approach to Monogenic Very Early Onset Inflammatory Bowel Disease.

Gastroenterology. 2014. doi: 10.1053/j.gastro.2014.07.023 [Epub ahead of print]

Uhlig HH, Schwerd T, Koletzko S, Shah N, Kammermeier J, Elkadri A, Ouahed J, Wilson DC, Travis S, Turner D, Klein C, Snapper SB, Muise AM.

Large B-Cell Lymphoma in an Adolescent Patient with IL-10 Receptor Deficiency and History of Infantile Inflammatory Bowel Disease.

Journal of Pediatric Gastroenterology and Nutrition. 2014. [Epub ahead of print]

Shouval DS, Ebens CL, Murchie R, McCann K, Rabah R, Klein C, Muise A, Snapper SB.

Inherited biallelic CSF3R mutations in severe congenital neutropenia.

Blood. 2014, 123(24),3811-7.

Triot A, Järvinen PM, Arostegui JI, Murugan D, Kohistani N, Dapena Díaz JL, Racek T, Puchalka J, Gertz EM, Schäffer AA, Kotlarz D, Pfeifer D, Díaz de Heredia Rubio C, Ozdemir MA, Patisroglu T, Karakukcu M, Sánchez de Toledo Codina J, Yagüe J, Touw I, Unal E, Klein C.

JAGN1 deficiency causes aberrant myeloid cell homeostasis and congenital neutropenia.

Nature Genetics. 2014. doi: 10.1038/ng.3069 [Epub ahead of print]

Boztug K, Järvinen PM, Salzer E, Racek T, Mönch S, Garncarz W, Gertz EM, Schäffer AA, Antonopoulos A, Haslam S, Ziesenitz L, Puchalka J, Diestelhorst J, Appaswamy G, Lescoeur B, Giambruno R, Bigenzahn JW, Elling U, Pfeifer D, Welte K, Brandes G, Sherkat R, van der Werff ten Bosch J, Rezaei N, Etzioni A, Bellanné-Chantelot C, Superti-Furga G, Penninger JM, Bennett KL, von Blume J, Dell A, Donadieu J, Klein C.

Jagunal-homolog 1 is a critical regulator of neutrophil function in fungal host defense.

Nature Genetics. 2014. doi: 10.1038/ng.3070 [Epub ahead of print]

Wirnsberger G, Zwolanek F, Stadlmann J, Tortola L, Liu SW, Järvinen P, Dürnberger G, Kozieradzki I, Sarao R, De Martinos A, Boztug K, Mechtler K, Kuchler K, Klein C, Elling U, Penninger JM.

A congenital neutrophil defect syndrome associated with mutations in VPS45.

New England Journal of Medicine. 2013, 369 (1),54-65.

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

IL-10R Polymorphisms Are Associated with Very-early-onset Ulcerative Colitis.

Inflammatory Bowel Disease. 2013, 19, 115-123.

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.

Clinical views and genetic analysis of patients with Wiskott-Aldrich syndrome: two novel mutations in the WASP gene.

Allergy. 2013, 68, 662-662.

Patiroglu T, Gungor EH, Ozdemir MA, Witzel M, Unal E, Karakurkcü M, Klein C.

Myeloperoxidase deficiency: the secret under the flag of unstained cell.

Turkish Journal of Haematology. 2013, 30, 232-233.

Patiroglu T, Eke Gungor H, Belohradsky JS, Unal E, Klein C

The phenotype of human STK4 deficiency.

Blood. 2012, 119, 3450-7.

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

Activating WASP mutations associated with X-linked neutropenia result in enhanced actin polymerization, altered cytoskeletal responses, and genomic instability in lymphocytes.

Journal of Experimental Medicine. 2010, 207, 1145-1152.

Westerberg LS, Meelu P, Babtista M, Adamovich DA, Cotta-de-Almeida V, See B, Rosen MK, Vandenberg P, Thrasher AJ, Klein C, Alt FW, Snapper SB.

Inflammatory Bowel Disease and Mutations Affecting the IL10 Receptor.

New England Journal of Medicine. 2009, 361, 2033-2045.

Glocker E, Kotlarz D, Boztug K, Gertz EM, Schäffer AA, Noyan F, Pero M, Diestelhorst J, Allroth A, Murugan D, Hätscher N, Pfeifer D, Sykora KW, Sauer M, Kreipe H, Lacher M, Nustede R, Woellner C, Baumann U, Salzer U, Koletzko S, Shah N, Segal A, Sauerbrey A, Buderus S, Snapper SB, Grimbacher B, Klein C.

Breakdown of T cell tolerance and autoimmunity in primary immunodeficiency.

Current Opinion in Immunology. 2008, 20, 646-54.

Westerberg LS, Klein C, Snapper SB.

The clinical, immunohematological, and molecular study of Iranian patients with severe congenital neutropenia.

Journal of Clinical Immunology. 2007, 27(5), 525-33.

Rezaei N, Moin M, Pourpak Z, Ramyar A, Izadyar M, Chavoshzadeh Z, Sherkat R, Aghamohammadi A, Yeganeh M, Mahmoudi M, Mahjoub F, Germeshausen M, Grudzien M, Horwitz MS, Klein C, Farhoudi A.

Lymphocyte dependent and Th2 cytokine associated colitis in mice deficient in Wiskott Aldrich Syndrome protein.

Gastroenterology. 2007, 133, 188-97.

Nguyen DD, Cotta de Almeida V, Maillard MH, Mizoguchi E, Klein C, Fuss I, Nagler C, Mizoguchi A, Bhan A, Snapper SB.

Retroviral WASP gene transfer into human hematopoietic stem cells reconstitutes the actin cytoskeleton in myeloid progeny cells differentiated in vitro.

Experimental Hematology. 2006, 34, 1162-1170.

Dewey RA, Avedillo Diez I, Ballmaier M, Filipovich A, Greil J, Gungör T, Happel CM, Maschan A, Noyan F, Pannicke U, Schwarz K, Snapper SB, Welte K, Klein C.

WASP-deficiency leads to global defects of directed leukocyte migration in vitro and in vivo.

Journal of Leukocyte Biology. 2005, 77,993-998.

Snapper SB, Meelu P, Nguyen D, Stockton B, Bozza P, Rosen F, Alt FW, von Andrian U, Klein C.

Gene Therapy for Wiskott Aldrich Syndrome - Rescue of T-Cell Signaling and Amelioration of Colitis upon Transplantation of Retrovirally Transduced Hematopoietic Stem Cells in Mice.

Blood. 2002, 101, 2159-2166.

Klein C, Nguyen D, Liu CH, Mizoguchi E, Bhan AK, Miki H, Takenawa T, Rosen FS, Alt FW, Mulligan RC, Snapper SB.

N-WASP deficiency reveals distinct pathways for cell surface projections and microbial actin-based motility.

Nature Cell Biology. 2001, 3, 897-904.

Snapper SB, Takeshima F, Anton I, Liu CH, Thomas SM, Nguyen D, Dudley D, Fraser H, Purich D, Lopez-Illasaca M, Klein C, Davidson L, Bronson R, Mulligan RC, Southwick F, Geha R, Goldberg MB, Rosen FS, Hartwig JH, Alt FW.