

# Holm H Uhlig

## List of Publications by Year in descending order

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Version: 2024-02-01

117  
papers

12,945  
citations

34493

54  
h-index

28425

109  
g-index

126  
all docs

126  
docs citations

126  
times ranked

20867  
citing authors

#	ARTICLE	IF	CITATIONS
1	Monogenic inflammatory bowel disease-genetic variants, functional mechanisms and personalised medicine in clinical practice. <i>Human Genetics</i> , 2023, 142, 599-611.	1.8	2
2	An Integrated Taxonomy for Monogenic Inflammatory Bowel Disease. <i>Gastroenterology</i> , 2022, 162, 859-876.	0.6	37
3	Valosin-containing protein-regulated endoplasmic reticulum stress causes NOD2-dependent inflammatory responses. <i>Scientific Reports</i> , 2022, 12, 3906.	1.6	0
4	Cessation of exclusive breastfeeding and seasonality, but not small intestinal bacterial overgrowth, are associated with environmental enteric dysfunction: A birth cohort study amongst infants in rural Kenya. <i>EClinicalMedicine</i> , 2022, 47, 101403.	3.2	3
5	UNC45A deficiency causes microvillus inclusion diseaseâ€‘like phenotype by impairing myosin VBâ€‘dependent apical trafficking. <i>Journal of Clinical Investigation</i> , 2022, 132, .	3.9	9
6	A method for the inference of cytokine interaction networks. <i>PLoS Computational Biology</i> , 2022, 18, e1010112.	1.5	2
7	Deconvolution of monocyte responses in inflammatory bowel disease reveals an IL-1 cytokine network that regulates IL-23 in genetic and acquired IL-10 resistance. <i>Gut</i> , 2021, 70, 1023-1036.	6.1	58
8	Predictive Prenatal Diagnosis for Infantileâ€‘onset Inflammatory Bowel Disease Because of Interleukinâ€‘10 Signalling Defects. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2021, 72, 276-281.	0.9	7
9	Gain-of-function variants in SYK cause immune dysregulation and systemic inflammation in humans and mice. <i>Nature Genetics</i> , 2021, 53, 500-510.	9.4	56
10	Inflammatory Bowel Disease in Patients with Congenital Chloride Diarrhoea. <i>Journal of Crohn's and Colitis</i> , 2021, 15, 1679-1685.	0.6	14
11	Deciphering the Transcriptomic Heterogeneity of Duodenal Coeliac Disease Biopsies. <i>International Journal of Molecular Sciences</i> , 2021, 22, 2551.	1.8	11
12	mTOR inhibitors reduce enteropathy, intestinal bleeding and colectomy rate in patients with juvenile polyposis of infancy with <i>PTEN-BMPRI1A</i> deletion. <i>Human Molecular Genetics</i> , 2021, 30, 1273-1282.	1.4	13
13	Human AGR2 Deficiency Causes Mucus Barrier Dysfunction and Infantile Inflammatory Bowel Disease. <i>Cellular and Molecular Gastroenterology and Hepatology</i> , 2021, 12, 1809-1830.	2.3	26
14	BCG Vaccineâ€‘Associated Complications in Patients with PTEN Hamartoma Tumor Syndrome. <i>Journal of Clinical Immunology</i> , 2021, 41, 1701-1705.	2.0	1
15	Biallelic <i>PI4KA</i> variants cause neurological, intestinal and immunological disease. <i>Brain</i> , 2021, 144, 3597-3610.	3.7	17
16	Functional and structural analysis of cytokine-selective IL6ST defects that cause recessive hyper-IgE syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 585-598.	1.5	20
17	Cells of the human intestinal tract mapped across space and time. <i>Nature</i> , 2021, 597, 250-255.	13.7	266
18	Inborn errors of IL-6 family cytokine responses. <i>Current Opinion in Immunology</i> , 2021, 72, 135-145.	2.4	25

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19	Congenital Diarrhea and Cholestatic Liver Disease: Phenotypic Spectrum Associated with MYO5B Mutations. <i>Journal of Clinical Medicine</i> , 2021, 10, 481.	1.0	20
20	Clinical Genomics for the Diagnosis of Monogenic Forms of Inflammatory Bowel Disease. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2021, 72, 456-473.	0.9	79
21	IL-1-driven stromal-neutrophil interactions define a subset of patients with inflammatory bowel disease that does not respond to therapies. <i>Nature Medicine</i> , 2021, 27, 1970-1981.	15.2	117
22	Paneth cell dysfunction and the intestinal microbiome in XIAP deficiency. <i>Science Immunology</i> , 2021, 6, eabm0293.	5.6	3
23	Remission of Inflammatory Bowel Disease in Glucose-6-Phosphatase 3 Deficiency by Allogeneic Haematopoietic Stem Cell Transplantation. <i>Journal of Crohn's and Colitis</i> , 2020, 14, 142-147.	0.6	27
24	Very Early Onset Inflammatory Bowel Disease: A Clinical Approach With a Focus on the Role of Genetics and Underlying Immune Deficiencies. <i>Inflammatory Bowel Diseases</i> , 2020, 26, 820-842.	0.9	100
25	Janus kinase inhibition for autoinflammation in patients with DNASE2 deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 145, 701-705.e8.	1.5	5
26	Demyelination After Anti-TNF Therapy: Who is at Risk?. <i>Journal of Crohn's and Colitis</i> , 2020, 14, 1651-1652.	0.6	3
27	Loss of IL-10 signaling in macrophages limits bacterial killing driven by prostaglandin E2. <i>Journal of Experimental Medicine</i> , 2020, 217, .	4.2	51
28	Dominant-negative mutations in human <i>IL6ST</i> underlie hyper-IgE syndrome. <i>Journal of Experimental Medicine</i> , 2020, 217, .	4.2	64
29	Absence of GP130 cytokine receptor signaling causes extended <i>Stx14</i> -Wiedemann syndrome. <i>Journal of Experimental Medicine</i> , 2020, 217, .	4.2	41
30	Alterations in T and B Cell Receptor Repertoires Patterns in Patients With IL10 Signaling Defects and History of Infantile-Onset IBD. <i>Frontiers in Immunology</i> , 2020, 11, 109.	2.2	11
31	A variant in <i>IL6ST</i> with a selective IL-11 signaling defect in human and mouse. <i>Bone Research</i> , 2020, 8, 24.	5.4	21
32	Discovery of CD80 and CD86 as recent activation markers on regulatory T cells by protein-RNA single-cell analysis. <i>Genome Medicine</i> , 2020, 12, 55.	3.6	61
33	Somatic mosaicism and common genetic variation contribute to the risk of very-early-onset inflammatory bowel disease. <i>Nature Communications</i> , 2020, 11, 995.	5.8	37
34	Prevalence and Clinical Features of Inflammatory Bowel Diseases Associated With Monogenic Variants, Identified by Whole-Exome Sequencing in 1000 Children at a Single Center. <i>Gastroenterology</i> , 2020, 158, 2208-2220.	0.6	81
35	Immune predictors of oral poliovirus vaccine immunogenicity among infants in South India. <i>Npj Vaccines</i> , 2020, 5, 27.	2.9	3
36	Corona Virus Disease 2019 and Paediatric Inflammatory Bowel Diseases. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2020, 70, 727-733.	0.9	114

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37	The Role of PTEN in Innate and Adaptive Immunity. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2019, 9, a036996.	2.9	24
38	The Short Chain Fatty Acid Butyrate Imprints an Antimicrobial Program in Macrophages. <i>Immunity</i> , 2019, 50, 432-445.e7.	6.6	612
39	Immunodeficiency, autoimmune thrombocytopenia and enterocolitis caused by autosomal recessive deficiency of <i>PIK3CD</i> -encoded phosphoinositide 3-kinase $\beta$ . <i>Haematologica</i> , 2019, 104, e483-e486.	1.7	26
40	Defining Faecal Calprotectin Thresholds as a Surrogate for Endoscopic and Histological Disease Activity in Ulcerative Colitis—a Prospective Analysis. <i>Journal of Crohn's and Colitis</i> , 2019, 13, 424-430.	0.6	54
41	Selective loss of function variants in <i>IL6ST</i> cause Hyper-IgE syndrome with distinct impairments of T-cell phenotype and function. <i>Haematologica</i> , 2019, 104, 609-621.	1.7	74
42	Mendelian Diseases and Inflammatory Bowel Disease—Data Mining for Genetic Risk and Disease-Associated Confounders. <i>Inflammatory Bowel Diseases</i> , 2018, 24, 467-470.	0.9	3
43	Human TGF- $\beta$ 1 deficiency causes severe inflammatory bowel disease and encephalopathy. <i>Nature Genetics</i> , 2018, 50, 344-348.	9.4	95
44	Antibody Concentrations Decrease 14-Fold in Children With Celiac Disease on a Gluten-Free Diet but Remain High at 3 Months. <i>Clinical Gastroenterology and Hepatology</i> , 2018, 16, 1442-1449.e5.	2.4	10
45	Translating Immunology into Therapeutic Concepts for Inflammatory Bowel Disease. <i>Annual Review of Immunology</i> , 2018, 36, 755-781.	9.5	121
46	Crohn's Disease in Niemann-Pick Disease Type C1: Caught in the Cross-Fire of Host-Microbial Interactions. <i>Digestive Diseases and Sciences</i> , 2018, 63, 811-813.	1.1	4
47	Interleukin-22 promotes phagolysosomal fusion to induce protection against <i>Salmonella enterica</i> Typhimurium in human epithelial cells. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 10118-10123.	3.3	33
48	A Spectrum of Genetic Variants Contributes to Immune Defects and Pathogenesis of Inflammatory Bowel Diseases. <i>Gastroenterology</i> , 2018, 154, 2022-2024.	0.6	7
49	Consequences of Identifying XIAP Deficiency in an Adult Patient With Inflammatory Bowel Disease. <i>Gastroenterology</i> , 2018, 155, 231-234.	0.6	22
50	Phenotypic and Genotypic Characterisation of Inflammatory Bowel Disease Presenting Before the Age of 2 years. <i>Journal of Crohn's and Colitis</i> , 2017, 11, 60-69.	0.6	146
51	Genome-wide association study implicates immune activation of multiple integrin genes in inflammatory bowel disease. <i>Nature Genetics</i> , 2017, 49, 256-261.	9.4	943
52	Exploring the genetic architecture of inflammatory bowel disease by whole-genome sequencing identifies association at ADCY7. <i>Nature Genetics</i> , 2017, 49, 186-192.	9.4	153
53	Impaired antibacterial autophagy links granulomatous intestinal inflammation in Niemann-Pick disease type C1 and XIAP deficiency with NOD2 variants in Crohn's disease. <i>Gut</i> , 2017, 66, 1060-1073.	6.1	126
54	Validation of Antibody-Based Strategies for Diagnosis of Pediatric Celiac Disease Without Biopsy. <i>Gastroenterology</i> , 2017, 153, 410-419.e17.	0.6	80

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55	BACH2 immunodeficiency illustrates an association between super-enhancers and haploinsufficiency. <i>Nature Immunology</i> , 2017, 18, 813-823.	7.0	113
56	Oral Vancomycin and Gentamicin for Treatment of Very Early Onset Inflammatory Bowel Disease. <i>Digestion</i> , 2017, 95, 310-313.	1.2	22
57	Vedolizumab in Paediatric Inflammatory Bowel Disease: A Retrospective Multi-Centre Experience From the Paediatric IBD Porto Group of ESPGHAN. <i>Journal of Crohn's and Colitis</i> , 2017, 11, 1230-1237.	0.6	82
58	Enhanced TH17 Responses in Patients with IL10 Receptor Deficiency and Infantile-onset IBD. <i>Inflammatory Bowel Diseases</i> , 2017, 23, 1950-1961.	0.9	28
59	Autophagy-Dependent Generation of Free Fatty Acids Is Critical for Normal Neutrophil Differentiation. <i>Immunity</i> , 2017, 47, 466-480.e5.	6.6	230
60	A biallelic mutation in <i>IL6ST</i> encoding the GP130 co-receptor causes immunodeficiency and craniosynostosis. <i>Journal of Experimental Medicine</i> , 2017, 214, 2547-2562.	4.2	158
61	Clinical Genomics in Inflammatory Bowel Disease. <i>Trends in Genetics</i> , 2017, 33, 629-641.	2.9	123
62	Circulating and Tissue-Resident CD4+ T Cells With Reactivity to Intestinal Microbiota Are Abundant in Healthy Individuals and Function Is Altered During Inflammation. <i>Gastroenterology</i> , 2017, 153, 1320-1337.e16.	0.6	246
63	Immune dysregulation in patients with PTEN hamartoma tumor syndrome: Analysis of FOXP3 regulatory T cells. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 607-620.e15.	1.5	77
64	Inherited GINS1 deficiency underlies growth retardation along with neutropenia and NK cell deficiency. <i>Journal of Clinical Investigation</i> , 2017, 127, 1991-2006.	3.9	115
65	From Genes to Mechanisms. <i>Inflammatory Bowel Diseases</i> , 2016, 22, 202-212.	0.9	58
66	The effect of azithromycin on the immunogenicity of oral poliovirus vaccine: a double-blind randomised placebo-controlled trial in seronegative Indian infants. <i>Lancet Infectious Diseases</i> , The, 2016, 16, 905-914.	4.6	55
67	Stem cell transplantation for tetratricopeptide repeat domain 7A deficiency: long-term follow-up. <i>Blood</i> , 2016, 128, 1306-1308.	0.6	40
68	Transplantation from a symptomatic carrier sister restores host defenses but does not prevent colitis in NEMO deficiency. <i>Clinical Immunology</i> , 2016, 164, 52-56.	1.4	38
69	Exclusive enteral nutrition in active pediatric Crohn disease: Effects on intestinal microbiota and immune regulation. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 592-596.	1.5	54
70	Variants in TRIM22 That Affect NOD2 Signaling Are Associated With Very-Early-Onset Inflammatory Bowel Disease. <i>Gastroenterology</i> , 2016, 150, 1196-1207.	0.6	88
71	A recessive form of extreme macrocephaly and mild intellectual disability complements the spectrum of PTEN hamartoma tumour syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 889-894.	1.4	6
72	ILC3 GM-CSF production and mobilisation orchestrate acute intestinal inflammation. <i>ELife</i> , 2016, 5, e10066.	2.8	185

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73	The Prevalence of Celiac Disease in Children and Adolescents in Germany. <i>Deutsches Arzteblatt International</i> , 2015, 112, 553-60.	0.6	58
74	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. <i>Nature Genetics</i> , 2015, 47, 717-726.	9.4	310
75	Defects in Nicotinamide-adenine Dinucleotide Phosphate Oxidase Genes NOX1 and DUOX2 in Very Early Onset Inflammatory Bowel Disease. <i>Cellular and Molecular Gastroenterology and Hepatology</i> , 2015, 1, 489-502.	2.3	127
76	Inflammatory Signaling by NOD-RIPK2 Is Inhibited by Clinically Relevant Type II Kinase Inhibitors. <i>Chemistry and Biology</i> , 2015, 22, 1174-1184.	6.2	101
77	Reduced sodium/proton exchanger NHE3 activity causes congenital sodium diarrhea. <i>Human Molecular Genetics</i> , 2015, 24, 6614-6623.	1.4	111
78	Sirolimus treatment of severe PTEN hamartoma tumor syndrome: case report and in vitro studies. <i>Pediatric Research</i> , 2014, 75, 527-534.	1.1	54
79	Toll-interacting Protein Modulates Colitis Susceptibility in Mice. <i>Inflammatory Bowel Diseases</i> , 2014, 20, 660-670.	0.9	28
80	The Diagnostic Approach to Monogenic Very Early Onset Inflammatory Bowel Disease. <i>Gastroenterology</i> , 2014, 147, 990-1007.e3.	0.6	559
81	Mutations in Tetratricopeptide Repeat Domain 7A Result in a Severe Form of Very Early Onset Inflammatory Bowel Disease. <i>Gastroenterology</i> , 2014, 146, 1028-1039.	0.6	175
82	Pulmonary Outcome in Former Preterm, Very Low Birth Weight Children with Bronchopulmonary Dysplasia: A Case-Control Follow-Up at School Age. <i>Journal of Pediatrics</i> , 2014, 164, 40-45.e4.	0.9	149
83	Characterization of Crohn disease in X-linked inhibitor of apoptosis-deficient male patients and female symptomatic carriers. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 134, 1131-1141.e9.	1.5	101
84	Targeted gene panel sequencing in children with very early onset inflammatory bowel disease: evaluation and prospective analysis. <i>Journal of Medical Genetics</i> , 2014, 51, 748-755.	1.5	91
85	Antibodies in the Diagnosis of Coeliac Disease: A Biopsy-Controlled, International, Multicentre Study of 376 Children with Coeliac Disease and 695 Controls. <i>PLoS ONE</i> , 2014, 9, e97853.	1.1	38
86	Monogenic diseases associated with intestinal inflammation: implications for the understanding of inflammatory bowel disease. <i>Gut</i> , 2013, 62, 1795-1805.	6.1	287
87	ILC1 Populations Join the Border Patrol. <i>Immunity</i> , 2013, 38, 630-632.	6.6	16
88	Early and nonreversible decrease of CD161 <sup>+</sup> /MAIT cells in HIV infection. <i>Blood</i> , 2013, 121, 951-961.	0.6	307
89	Determination of IgG and IgA Antibodies Against Native Gliadin Is Not Helpful for the Diagnosis of Coeliac Disease in Children Up to 2 Years Old. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2012, 55, 21-25.	0.9	11
90	Lymphoid microenvironments and innate lymphoid cells in the gut. <i>Trends in Immunology</i> , 2012, 33, 289-296.	2.9	119

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91	Autoimmunity, Intestinal Lymphoid Hyperplasia, and Defects in Mucosal B-Cell Homeostasis in Patients With PTEN Hamartoma Tumor Syndrome. <i>Gastroenterology</i> , 2012, 142, 1093-1096.e6.	0.6	61
92	Recurrent Abdominal Pain in Childhood. <i>Deutsches A&amp;#x0308;rzteblatt International</i> , 2011, 108, 295-304.	0.6	49
93	Use of likelihood ratios improves clinical interpretation of IgG and IgA anti-DGP antibody testing for celiac disease in adults and children. <i>Clinical Biochemistry</i> , 2011, 44, 248-250.	0.8	9
94	Innate lymphoid cells drive interleukin-23-dependent innate intestinal pathology. <i>Nature</i> , 2010, 464, 1371-1375.	13.7	978
95	NOD2 mutations predict the risk for surgery in pediatric-onset Crohn's disease. <i>Journal of Pediatric Surgery</i> , 2010, 45, 1591-1597.	0.8	36
96	Mouse models of intestinal inflammation as tools to understand the pathogenesis of inflammatory bowel disease. <i>European Journal of Immunology</i> , 2009, 39, 2021-2026.	1.6	42
97	Frequency of indeterminate colitis in children and adults with IBD â€” a metaanalysis. <i>Journal of Crohn's and Colitis</i> , 2009, 3, 277-281.	0.6	124
98	Antibodies Against Deamidated Gliadin as New and Accurate Biomarkers of Childhood Coeliac Disease. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2009, 49, 52-58.	0.9	59
99	Animal Models of Intestinal Inflammation: Clues to the Pathogenesis of Inflammatory Bowel Disease. <i>Novartis Foundation Symposium</i> , 2008, , 164-178.	1.2	18
100	Control of Immune Pathology by Regulatory T Cells. <i>Novartis Foundation Symposium</i> , 2008, , 92-105.	1.2	96
101	Homing of Intestinal Immune Cells. <i>Novartis Foundation Symposium</i> , 2008, 263, 179-192.	1.2	9
102	Segmented filamentous bacteria in a defined bacterial cocktail induce intestinal inflammation in SCID mice reconstituted with CD45RB <sup>high</sup> CD4 <sup>+</sup> T cells. <i>Inflammatory Bowel Diseases</i> , 2007, 13, 1202-1211.	0.9	177
103	Differential Activity of IL-12 and IL-23 in Mucosal and Systemic Innate Immune Pathology. <i>Immunity</i> , 2006, 25, 309-318.	6.6	615
104	Control of Intestinal Inflammation by Regulatory T Cells. <i>Inflammatory Bowel Diseases</i> , 2006, 12, S4-S5.	0.9	0
105	Characterization of Foxp3 <sup>+</sup> CD4 <sup>+</sup> CD25 <sup>+</sup> and IL-10-Secreting CD4 <sup>+</sup> CD25 <sup>+</sup> T Cells during Cure of Colitis. <i>Journal of Immunology</i> , 2006, 177, 5852-5860.	0.4	404
106	Regression of Pancreatic Diabetes in Chronic Hereditary Pancreatitis. <i>Diabetes Care</i> , 2006, 29, 1981-1982.	4.3	7
107	Involvement of Innate Immunity in the Development of Inflammatory and Autoimmune Diseases. <i>Annals of the New York Academy of Sciences</i> , 2005, 1051, 787-798.	1.8	76
108	Regulatory T cells and intestinal homeostasis. <i>Immunological Reviews</i> , 2005, 204, 184-194.	2.8	255

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109	The role of mucosal T lymphocytes in regulating intestinal inflammation. <i>Seminars in Immunopathology</i> , 2005, 27, 167-180.	4.0	23
110	Essential role for CD103 in the T cell-mediated regulation of experimental colitis. <i>Journal of Experimental Medicine</i> , 2005, 202, 1051-1061.	4.2	450
111	Animal models of intestinal inflammation: clues to the pathogenesis of inflammatory bowel disease. <i>Novartis Foundation Symposium</i> , 2004, 263, 164-74; discussion 174-8, 211-8.	1.2	13
112	Cutting Edge: Cure of Colitis by CD4+CD25+ Regulatory T Cells. <i>Journal of Immunology</i> , 2003, 170, 3939-3943.	0.4	858
113	Dendritic cells and the intestinal bacterial flora: a role for localized mucosal immune responses. <i>Journal of Clinical Investigation</i> , 2003, 112, 648-651.	3.9	79
114	Serological differentiation of inflammatory bowel diseases. <i>European Journal of Gastroenterology and Hepatology</i> , 2002, 14, 129-135.	0.8	43
115	Antibody response to dietary and autoantigens in $\text{G}\mu\text{i}2$ -deficient mice. <i>European Journal of Gastroenterology and Hepatology</i> , 2001, 13, 1421-1429.	0.8	10
116	A monoclonal antibody that recognizes a potential coeliac-toxic repetitive pentapeptide epitope in gliadins. <i>European Journal of Gastroenterology and Hepatology</i> , 2001, 13, 1189-1193.	0.8	122
117	Use of the phage display technique for detection of epitopes recognized by polyclonal rabbit gliadin antibodies. <i>FEBS Letters</i> , 1998, 433, 103-107.	1.3	19