Aleixo M Muise

List of Publications by Year in Descending Order

111 papers	5,430 citations	41 h-index	72 g-index
120 ext. papers	6,683 ext. citations	9 avg, IF	5.33 L-index

#	Paper	IF	Citations
111	A Machine Learning Approach to Identifying Causal Monogenic Variants in Inflammatory Bowel Disease **2022**, 1, 171-179		
110	Valosin-containing protein-regulated endoplasmic reticulum stress causes NOD2-dependent inflammatory responses.. *Scientific Reports*, **2022**, 12, 3906	4.9	
109	Deficiency in X-linked inhibitor of apoptosis protein promotes susceptibility to microbial triggers of intestinal inflammation. *Science Immunology*, **2021**, 6, eabf7473	28	2
108	An integrated taxonomy for monogenic inflammatory bowel disease. *Gastroenterology*, **2021**,	13.3	4
107	ARPC1B binds WASP to control actin polymerization and curtail tonic signaling in B cells. *JCI Insight*, **2021**, 6,	9.9	1
106	Multisystem Autoimmune Inflammatory Disease, Including Colitis, Due to Inborn Error of Immunity. *Pediatrics*, **2021**, 148,	7.4	1
105	Clinical Genomics for the Diagnosis of Monogenic Forms of Inflammatory Bowel Disease: A Position Paper From the Paediatric IBD Porto Group of European Society of Paediatric Gastroenterology, Hepatology and Nutrition. *Journal of Pediatric Gastroenterology and Nutrition*, **2021**, 72, 456-473	2.8	19
104	A Systematic Review of Monogenic Inflammatory Bowel Disease. *Clinical Gastroenterology and Hepatology*, **2021**,	6.9	5
103	Novel CARMIL2 loss-of-function variants are associated with pediatric inflammatory bowel disease. *Scientific Reports*, **2021**, 11, 5945	4.9	5
102	Variants in STXBP3 are Associated with Very Early Onset Inflammatory Bowel Disease, Bilateral Sensorineural Hearing Loss and Immune Dysregulation. *Journal of Crohn's and Colitis*, **2021**, 15, 1908-1919	1.5	0
101	Human autoinflammatory disease reveals ELF4 as a transcriptional regulator of inflammation. *Nature Immunology*, **2021**, 22, 1118-1126	19.1	3
100	Natural History of Very Early Onset Inflammatory Bowel Disease in North America: A Retrospective Cohort Study. *Inflammatory Bowel Diseases*, **2021**, 27, 295-302	4.5	13
99	Diagnostic Delay Is Associated With Complicated Disease and Growth Impairment in Paediatric Crohn's Disease. *Journal of Crohn's and Colitis*, **2021**, 15, 419-431	1.5	6
98	Cutting Edge: NOX2 NADPH Oxidase Controls Infection by an Intracellular Bacterial Pathogen through Limiting the Type 1 IFN Response. *Journal of Immunology*, **2021**, 206, 323-328	5.3	3
97	Whipple disease mimicking inflammatory bowel disease. *Intestinal Research*, **2021**, 19, 119-125	4.1	1
96	Predictive Prenatal Diagnosis for Infantile-onset Inflammatory Bowel Disease Because of Interleukin-10 Signalling Defects. *Journal of Pediatric Gastroenterology and Nutrition*, **2021**, 72, 276-281	2.8	3
95	Mutation spectrum of NOD2 reveals recessive inheritance as a main driver of Early Onset Crohn's Disease. *Scientific Reports*, **2021**, 11, 5595	4.9	11

94	Gain-of-function variants in SYK cause immune dysregulation and systemic inflammation in humans and mice. *Nature Genetics*, **2021**, 53, 500-510	36.3	11
93	2019 Harry Shwachman Award: Dr Anne M. Griffiths, MD, FRCPC. *Journal of Pediatric Gastroenterology and Nutrition*, **2020**, 70, 405	2.8	
92	NOX1 Regulates Collective and Planktonic Cell Migration: Insights From Patients With Pediatric-Onset IBD and NOX1 Deficiency. *Inflammatory Bowel Diseases*, **2020**, 26, 1166-1176	4.5	4
91	Somatic mosaicism and common genetic variation contribute to the risk of very-early-onset inflammatory bowel disease. *Nature Communications*, **2020**, 11, 995	17.4	21
90	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**, 158, 2208-2220	13.3	32
89	Drug Screen Identifies Leflunomide for Treatment of Inflammatory Bowel Disease Caused by TTC7A Deficiency. *Gastroenterology*, **2020**, 158, 1000-1015	13.3	16
88	Very Early Onset Inflammatory Bowel Disease: A Clinical Approach With a Focus on the Role of Genetics and Underlying Immune Deficiencies. *Inflammatory Bowel Diseases*, **2020**, 26, 820-842	4.5	40
87	Utilization of Whole Exome Sequencing Data to Identify Clinically Relevant Pharmacogenomic Variants in Pediatric Inflammatory Bowel Disease. *Clinical and Translational Gastroenterology*, **2020**, 11, e00263	4.2	1
86	The E3 ubiquitin ligase UBR5 interacts with TTC7A and may be associated with very early onset inflammatory bowel disease. *Scientific Reports*, **2020**, 10, 18648	4.9	2
85	Pediatric Diarrheal Disorders **2020**, 143-157		
84	Advanced Understanding of Monogenic Inflammatory Bowel Disease. *Frontiers in Pediatrics*, **2020**, 8, 618918	3.4	3
83	Palmitoylation of NOD1 and NOD2 is required for bacterial sensing. *Science*, **2019**, 366, 460-467	33.3	45
82	CARMIL2 Deficiency Presenting as Very Early Onset Inflammatory Bowel Disease. *Inflammatory Bowel Diseases*, **2019**, 25, 1788-1795	4.5	16
81	Very Early Onset Inflammatory Bowel Disease (VEOIBD) **2019**, 383-404		
80	Intensified Infliximab Induction is Associated with Improved Response and Decreased Colectomy in Steroid-Refractory Paediatric Ulcerative Colitis. *Journal of Crohn's and Colitis*, **2019**, 13, 982-989	1.5	14
79	Novel Exonic Deletions in TTC7A in a Newborn with Multiple Intestinal Atresia and Combined Immunodeficiency. *Journal of Clinical Immunology*, **2019**, 39, 616-619	5.7	3
78	Sequencing and Mapping IBD Genes to Individual Causative Variants and Their Clinical Relevance **2019**, 117-139		
77	Application of Whole Exome Sequencing in Congenital Secretory Diarrhea Diagnosis. *Journal of Pediatric Gastroenterology and Nutrition*, **2019**, 68, e106-e108	2.8	1

76	Human RIPK1 deficiency causes combined immunodeficiency and inflammatory bowel diseases. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 970-975	11.5	79
75	TTC7A: Steward of Intestinal Health. *Cellular and Molecular Gastroenterology and Hepatology*, **2019**, 7, 555-570	7.9	29
74	Intestinal Inflammation and Dysregulated Immunity in Patients With Inherited Caspase-8 Deficiency. *Gastroenterology*, **2019**, 156, 275-278	13.3	61
73	Human TGF-β deficiency causes severe inflammatory bowel disease and encephalopathy. *Nature Genetics*, **2018**, 50, 344-348	36.3	67
72	Advances in Evaluation of Chronic Diarrhea in Infants. *Gastroenterology*, **2018**, 154, 2045-2059.e6	13.3	81
71	Human ALPI deficiency causes inflammatory bowel disease and highlights a key mechanism of gut homeostasis. *EMBO Molecular Medicine*, **2018**, 10,	12	33
70	Long-term outcomes for children with very early-onset colitis: Implications for surgical management. *Journal of Pediatric Surgery*, **2018**, 53, 964-967	2.6	14
69	Inflammatory Bowel Disease in Primary Immunodeficiencies **2018**, 167-181		
68	Monogenic Intestinal Epithelium Defects and the Development of Inflammatory Bowel Disease. *Physiology*, **2018**, 33, 360-369	9.8	8
67	Inherited p40phox deficiency differs from classic chronic granulomatous disease. *Journal of Clinical Investigation*, **2018**, 128, 3957-3975	15.9	65
66	Diagnostic delay in Canadian children with inflammatory bowel disease is more common in Crohn's disease and associated with decreased height. *Archives of Disease in Childhood*, **2018**, 103, 319-326	2.2	23
65	An ATG16L1-dependent pathway promotes plasma membrane repair and limits Listeria monocytogenes cell-to-cell spread. *Nature Microbiology*, **2018**, 3, 1472-1485	26.6	40
64	Inflammatory Bowel Disease: What Very Early Onset Disease Teaches Us. *Gastroenterology Clinics of North America*, **2018**, 47, 755-772	4.4	25
63	Very early onset IBD: novel genetic aetiologies. *Current Opinion in Allergy and Clinical Immunology*, **2018**, 18, 470-480	3.3	12
62	Loss of the Arp2/3 complex component ARPC1B causes platelet abnormalities and predisposes to inflammatory disease. *Nature Communications*, **2017**, 8, 14816	17.4	128
61	Ankyrin repeat and zinc-finger domain-containing 1 mutations are associated with infantile-onset inflammatory bowel disease. *Journal of Biological Chemistry*, **2017**, 292, 7904-7920	5.4	15
60	Enhanced TH17 Responses in Patients with IL10 Receptor Deficiency and Infantile-onset IBD. *Inflammatory Bowel Diseases*, **2017**, 23, 1950-1961	4.5	19
59	Clinical Genomics in Inflammatory Bowel Disease. *Trends in Genetics*, **2017**, 33, 629-641	8.5	91

58	Variants in TRIM22 That Affect NOD2 Signaling Are Associated With Very-Early-Onset Inflammatory Bowel Disease. *Gastroenterology*, **2016**, 150, 1196-1207	13.3	69
57	Multilabel immunofluorescence and antigen reprobing on formalin-fixed paraffin-embedded sections: novel applications for precision pathology diagnosis. *Modern Pathology*, **2016**, 29, 557-69	9.8	16
56	Large B-Cell Lymphoma in an Adolescent Patient With Interleukin-10 Receptor Deficiency and History of Infantile Inflammatory Bowel Disease. *Journal of Pediatric Gastroenterology and Nutrition* , **2016**, 63, e15-7	2.8	26
55	A Frameshift in CSF2RB Predominant Among Ashkenazi Jews Increases Risk for Crohn's Disease and Reduces Monocyte Signaling via GM-CSF. *Gastroenterology*, **2016**, 151, 710-723.e2	13.3	40
54	Mucosa-Associated Ileal Microbiota in New-Onset Pediatric Crohn's Disease. *Inflammatory Bowel Diseases*, **2016**, 22, 1533-9	4.5	32
53	Interleukin 1Mediates Intestinal Inflammation in Mice and Patients With Interleukin 10 Receptor Deficiency. *Gastroenterology*, **2016**, 151, 1100-1104	13.3	102
52	The diaphanous-related formins promote protrusion formation and cell-to-cell spread of Listeria monocytogenes. *Journal of Infectious Diseases*, **2015**, 211, 1185-95	7	36
51	Unrelated donor hematopoietic stem cell transplantation for infantile enteropathy due to IL-10/IL-10 receptor defect. *Pediatric Transplantation*, **2015**, 19, E101-3	1.8	7
50	Defects in NADPH Oxidase Genes and in Very Early Onset Inflammatory Bowel Disease. *Cellular and Molecular Gastroenterology and Hepatology*, **2015**, 1, 489-502	7.9	91
49	Reduced sodium/proton exchanger NHE3 activity causes congenital sodium diarrhea. *Human Molecular Genetics*, **2015**, 24, 6614-23	5.6	80
48	Mutations in Plasmalemma Vesicle Associated Protein Result in Sieving Protein-Losing Enteropathy Characterized by Hypoproteinemia, Hypoalbuminemia, and Hypertriglyceridemia. *Cellular and Molecular Gastroenterology and Hepatology*, **2015**, 1, 381-394.e7	7.9	32
47	Fatal autoimmunity in mice reconstituted with human hematopoietic stem cells encoding defective FOXP3. *Blood*, **2015**, 125, 3886-95	2.2	26
46	Rac1 Polymorphisms and Thiopurine Efficacy in Children With Inflammatory Bowel Disease. *Journal of Pediatric Gastroenterology and Nutrition*, **2015**, 61, 404-7	2.8	8
45	Very early-onset inflammatory bowel disease: gaining insight through focused discovery. *Inflammatory Bowel Diseases*, **2015**, 21, 1166-75	4.5	67
44	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	5.7	50
43	Interleukin-10 receptor signaling in innate immune cells regulates mucosal immune tolerance and anti-inflammatory macrophage function. *Immunity*, **2014**, 40, 706-19	32.3	337
42	Listeria monocytogenes exploits efferocytosis to promote cell-to-cell spread. *Nature*, **2014**, 509, 230-4	50.4	96
41	Incidence, outcomes, and health services burden of very early onset inflammatory bowel disease. *Gastroenterology*, **2014**, 147, 803-813.e7; quiz e14-5	13.3	162

40	Variants in nicotinamide adenine dinucleotide phosphate oxidase complex components determine susceptibility to very early onset inflammatory bowel disease. *Gastroenterology*, **2014**, 147, 680-689.e2	13.3	88
39	Interleukin 10 receptor signaling: master regulator of intestinal mucosal homeostasis in mice and humans. *Advances in Immunology*, **2014**, 122, 177-210	5.6	175
38	Novel de novo mutations of the interleukin-10 receptor gene lead to infantile onset inflammatory bowel disease. *Journal of Crohn's and Colitis*, **2014**, 8, 1551-6	1.5	24
37	A CARD9 polymorphism is associated with decreased likelihood of persistent conjugated hyperbilirubinemia in intestinal failure. *PLoS ONE*, **2014**, 9, e85915	3.7	9
36	Higher activity of the inducible nitric oxide synthase contributes to very early onset inflammatory bowel disease. *Clinical and Translational Gastroenterology*, **2014**, 5, e46	4.2	55
35	Protein tyrosine phosphatase 1 targets apical junction complex proteins in the intestine and regulates epithelial permeability. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 693-8	11.5	48
34	Microvillus inclusion disease: loss of Myosin vb disrupts intracellular traffic and cell polarity. *Traffic*, **2014**, 15, 22-42	5.7	45
33	A novel nonsense mutation in the EpCAM gene in a patient with congenital tufting enteropathy. *Journal of Pediatric Gastroenterology and Nutrition*, **2014**, 58, 18-21	2.8	17
32	The diagnostic approach to monogenic very early onset inflammatory bowel disease. *Gastroenterology*, **2014**, 147, 990-1007.e3	13.3	422
31	Infliximab maintains durable response and facilitates catch-up growth in luminal pediatric Crohn's disease. *Inflammatory Bowel Diseases*, **2014**, 20, 1177-86	4.5	67
30	Mutations in tetratricopeptide repeat domain 7A result in a severe form of very early onset inflammatory bowel disease. *Gastroenterology*, **2014**, 146, 1028-39	13.3	138
29	IL-10R polymorphisms are associated with very-early-onset ulcerative colitis. *Inflammatory Bowel Diseases*, **2013**, 19, 115-23	4.5	178
28	Clinical outcome in IL-10- and IL-10 receptor-deficient patients with or without hematopoietic stem cell transplantation. *Journal of Allergy and Clinical Immunology*, **2013**, 131, 825-30	11.5	195
27	Interleukin-6 is associated with steroid resistance and reflects disease activity in severe pediatric ulcerative colitis. *Journal of Crohn's and Colitis*, **2013**, 7, 916-22	1.5	35
26	The NOD2insC polymorphism is associated with worse outcome following ileal pouch-anal anastomosis for ulcerative colitis. *Gut*, **2013**, 62, 1433-9	19.2	72
25	Association between a multi-locus genetic risk score and inflammatory bowel disease. *Bioinformatics and Biology Insights*, **2013**, 7, 143-52	5.3	7
24	Host and bacterial factors that regulate LC3 recruitment to Listeria monocytogenes during the early stages of macrophage infection. *Autophagy*, **2013**, 9, 985-95	10.2	91
23	Anti-TNF, infliximab, and adalimumab can be effective in eosinophilic bowel disease. *Journal of Pediatric Gastroenterology and Nutrition*, **2013**, 56, 492-7	2.8	21

22	Infliximab-induced psoriasis and psoriasiform skin lesions in pediatric Crohn disease and a potential association with IL-23 receptor polymorphisms. *Journal of Pediatric Gastroenterology and Nutrition*, **2013**, 56, 512-8	2.8	45
21	Rac2-deficiency leads to exacerbated and protracted colitis in response to Citrobacter rodentium infection. *PLoS ONE*, **2013**, 8, e61629	3.7	19
20	The Authors' reply: Figure 1. *Gut*, **2012**, 61, 1097.2-1098	19.2	
19	NADPH oxidase complex and IBD candidate gene studies: identification of a rare variant in NCF2 that results in reduced binding to RAC2. *Gut*, **2012**, 61, 1028-35	19.2	129
18	Single nucleotide polymorphisms that increase expression of the guanosine triphosphatase RAC1 are associated with ulcerative colitis. *Gastroenterology*, **2011**, 141, 633-41	13.3	58
17	C-reactive protein (CRP), erythrocyte sedimentation rate (ESR) or both? A systematic evaluation in pediatric ulcerative colitis. *Journal of Crohns and Colitis*, **2011**, 5, 423-9	1.5	48
16	Listeriolysin O suppresses phospholipase C-mediated activation of the microbicidal NADPH oxidase to promote Listeria monocytogenes infection. *Cell Host and Microbe*, **2011**, 10, 627-34	23.4	60
15	Replication of genetic variation in the MYO9B gene in Crohn's disease. *Human Immunology*, **2011**, 72, 592-7	2.3	15
14	NADPH oxidase complex and IBD Candidate Gene studiesO-18.. *Inflammatory Bowel Diseases*, **2011**, 17, S8	4.5	
13	Common variants at five new loci associated with early-onset inflammatory bowel disease. *Nature Genetics*, **2009**, 41, 1335-40	36.3	389
12	Apical junction complex proteins and ulcerative colitis: a focus on the PTPRS gene. *Expert Review of Molecular Diagnostics*, **2008**, 8, 465-77	3.8	7
11	Protein-tyrosine phosphatase sigma is associated with ulcerative colitis. *Current Biology*, **2007**, 17, 1212-8	8.3	46
10	Low levels of procalcitonin during episodes of necrotizing enterocolitis. *Digestive Diseases and Sciences*, **2007**, 52, 2972-6	4	17
9	Biliary atresia with choledochal cyst: implications for classification. *Clinical Gastroenterology and Hepatology*, **2006**, 4, 1411-4	6.9	29
8	Eating disorders in adolescent boys: a review of the adolescent and young adult literature. *Journal of Adolescent Health*, **2003**, 33, 427-35	5.8	99
7	Are children with Kawasaki disease and prolonged fever at risk for macrophage activation syndrome?. *Pediatrics*, **2003**, 112, e495	7.4	40
6	Regulation of adipogenesis by a transcriptional repressor that modulates MAPK activation. *Journal of Biological Chemistry*, **2001**, 276, 10199-206	5.4	88
5	The risk of myocardial infarction in HIV-infected patients receiving HAART: a case report. *International Journal of STD and AIDS*, **2001**, 12, 612-3	1.4	5

4	Transcriptional regulation by the gamma5 subunit of a heterotrimeric G protein during adipogenesis. *EMBO Journal*, **1999**, 18, 4004-12	13	51
3	Enzymic characterization of a novel member of the regulatory B-like carboxypeptidase with transcriptional repression function: stimulation of enzymic activity by its target DNA. *Biochemical Journal*, **1999**, 343, 341-345	3.8	20
2	A eukaryotic transcriptional repressor with carboxypeptidase activity. *Nature*, **1995**, 378, 92-6	50.4	146
1	Mutation spectrum ofNOD2reveals recessive inheritance as a main driver of Early Onset Crohn's Disease		2