

# Aleixo M Muise

## List of Publications by Citations

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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	The diagnostic approach to monogenic very early onset inflammatory bowel disease. <i>Gastroenterology</i> , <b>2014</b> , 147, 990-1007.e3	13.3	422
110	Common variants at five new loci associated with early-onset inflammatory bowel disease. <i>Nature Genetics</i> , <b>2009</b> , 41, 1335-40	36.3	389
109	Interleukin-10 receptor signaling in innate immune cells regulates mucosal immune tolerance and anti-inflammatory macrophage function. <i>Immunity</i> , <b>2014</b> , 40, 706-19	32.3	337
108	Clinical outcome in IL-10- and IL-10 receptor-deficient patients with or without hematopoietic stem cell transplantation. <i>Journal of Allergy and Clinical Immunology</i> , <b>2013</b> , 131, 825-30	11.5	195
107	IL-10R polymorphisms are associated with very-early-onset ulcerative colitis. <i>Inflammatory Bowel Diseases</i> , <b>2013</b> , 19, 115-23	4.5	178
106	Interleukin 10 receptor signaling: master regulator of intestinal mucosal homeostasis in mice and humans. <i>Advances in Immunology</i> , <b>2014</b> , 122, 177-210	5.6	175
105	Incidence, outcomes, and health services burden of very early onset inflammatory bowel disease. <i>Gastroenterology</i> , <b>2014</b> , 147, 803-813.e7; quiz e14-5	13.3	162
104	A eukaryotic transcriptional repressor with carboxypeptidase activity. <i>Nature</i> , <b>1995</b> , 378, 92-6	50.4	146
103	Mutations in tetratricopeptide repeat domain 7A result in a severe form of very early onset inflammatory bowel disease. <i>Gastroenterology</i> , <b>2014</b> , 146, 1028-39	13.3	138
102	NADPH oxidase complex and IBD candidate gene studies: identification of a rare variant in NCF2 that results in reduced binding to RAC2. <i>Gut</i> , <b>2012</b> , 61, 1028-35	19.2	129
101	Loss of the Arp2/3 complex component ARPC1B causes platelet abnormalities and predisposes to inflammatory disease. <i>Nature Communications</i> , <b>2017</b> , 8, 14816	17.4	128
100	Interleukin 10 Mediates Intestinal Inflammation in Mice and Patients With Interleukin 10 Receptor Deficiency. <i>Gastroenterology</i> , <b>2016</b> , 151, 1100-1104	13.3	102
99	Eating disorders in adolescent boys: a review of the adolescent and young adult literature. <i>Journal of Adolescent Health</i> , <b>2003</b> , 33, 427-35	5.8	99
98	Listeria monocytogenes exploits efferocytosis to promote cell-to-cell spread. <i>Nature</i> , <b>2014</b> , 509, 230-4	50.4	96
97	Defects in NADPH Oxidase Genes and in Very Early Onset Inflammatory Bowel Disease. <i>Cellular and Molecular Gastroenterology and Hepatology</i> , <b>2015</b> , 1, 489-502	7.9	91
96	Clinical Genomics in Inflammatory Bowel Disease. <i>Trends in Genetics</i> , <b>2017</b> , 33, 629-641	8.5	91
95	Host and bacterial factors that regulate LC3 recruitment to Listeria monocytogenes during the early stages of macrophage infection. <i>Autophagy</i> , <b>2013</b> , 9, 985-95	10.2	91

94	Variants in nicotinamide adenine dinucleotide phosphate oxidase complex components determine susceptibility to very early onset inflammatory bowel disease. <i>Gastroenterology</i> , <b>2014</b> , 147, 680-689.e2	13.3	88
93	Regulation of adipogenesis by a transcriptional repressor that modulates MAPK activation. <i>Journal of Biological Chemistry</i> , <b>2001</b> , 276, 10199-206	5.4	88
92	Advances in Evaluation of Chronic Diarrhea in Infants. <i>Gastroenterology</i> , <b>2018</b> , 154, 2045-2059.e6	13.3	81
91	Reduced sodium/proton exchanger NHE3 activity causes congenital sodium diarrhea. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 6614-23	5.6	80
90	Human RIPK1 deficiency causes combined immunodeficiency and inflammatory bowel diseases. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2019</b> , 116, 970-975	11.5	79
89	The NOD2insC polymorphism is associated with worse outcome following ileal pouch-anal anastomosis for ulcerative colitis. <i>Cut</i> , <b>2013</b> , 62, 1433-9	19.2	72
88	Variants in TRIM22 That Affect NOD2 Signaling Are Associated With Very-Early-Onset Inflammatory Bowel Disease. <i>Gastroenterology</i> , <b>2016</b> , 150, 1196-1207	13.3	69
87	Human TGF- $\beta$ 1 deficiency causes severe inflammatory bowel disease and encephalopathy. <i>Nature Genetics</i> , <b>2018</b> , 50, 344-348	36.3	67
86	Very early-onset inflammatory bowel disease: gaining insight through focused discovery. <i>Inflammatory Bowel Diseases</i> , <b>2015</b> , 21, 1166-75	4.5	67
85	Infliximab maintains durable response and facilitates catch-up growth in luminal pediatric Crohn's disease. <i>Inflammatory Bowel Diseases</i> , <b>2014</b> , 20, 1177-86	4.5	67
84	Inherited p40phox deficiency differs from classic chronic granulomatous disease. <i>Journal of Clinical Investigation</i> , <b>2018</b> , 128, 3957-3975	15.9	65
83	Intestinal Inflammation and Dysregulated Immunity in Patients With Inherited Caspase-8 Deficiency. <i>Gastroenterology</i> , <b>2019</b> , 156, 275-278	13.3	61
82	Listeriolysin O suppresses phospholipase C-mediated activation of the microbicidal NADPH oxidase to promote <i>Listeria monocytogenes</i> infection. <i>Cell Host and Microbe</i> , <b>2011</b> , 10, 627-34	23.4	60
81	Single nucleotide polymorphisms that increase expression of the guanosine triphosphatase RAC1 are associated with ulcerative colitis. <i>Gastroenterology</i> , <b>2011</b> , 141, 633-41	13.3	58
80	Higher activity of the inducible nitric oxide synthase contributes to very early onset inflammatory bowel disease. <i>Clinical and Translational Gastroenterology</i> , <b>2014</b> , 5, e46	4.2	55
79	Transcriptional regulation by the gamma5 subunit of a heterotrimeric G protein during adipogenesis. <i>EMBO Journal</i> , <b>1999</b> , 18, 4004-12	13	51
78	Very early onset inflammatory bowel disease associated with aberrant trafficking of IL-10R1 and cure by T cell replete haploidentical bone marrow transplantation. <i>Journal of Clinical Immunology</i> , <b>2014</b> , 34, 331-9	5.7	50
77	Protein tyrosine phosphatase $\zeta$ targets apical junction complex proteins in the intestine and regulates epithelial permeability. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2014</b> , 111, 693-8	11.5	48

76	C-reactive protein (CRP), erythrocyte sedimentation rate (ESR) or both? A systematic evaluation in pediatric ulcerative colitis. <i>Journal of Crohns and Colitis</i> , <b>2011</b> , 5, 423-9	1.5	48
75	Protein-tyrosine phosphatase sigma is associated with ulcerative colitis. <i>Current Biology</i> , <b>2007</b> , 17, 1212-8.3	4.6	46
74	Palmitoylation of NOD1 and NOD2 is required for bacterial sensing. <i>Science</i> , <b>2019</b> , 366, 460-467	33.3	45
73	Microvillus inclusion disease: loss of Myosin vb disrupts intracellular traffic and cell polarity. <i>Traffic</i> , <b>2014</b> , 15, 22-42	5.7	45
72	Infliximab-induced psoriasis and psoriasiform skin lesions in pediatric Crohn disease and a potential association with IL-23 receptor polymorphisms. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2013</b> , 56, 512-8	2.8	45
71	Are children with Kawasaki disease and prolonged fever at risk for macrophage activation syndrome?. <i>Pediatrics</i> , <b>2003</b> , 112, e495	7.4	40
70	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> , <b>2020</b> , 26, 820-842	4.5	40
69	A Frameshift in CSF2RB Predominant Among Ashkenazi Jews Increases Risk for Crohn's Disease and Reduces Monocyte Signaling via GM-CSF. <i>Gastroenterology</i> , <b>2016</b> , 151, 710-723.e2	13.3	40
68	An ATG16L1-dependent pathway promotes plasma membrane repair and limits Listeria monocytogenes cell-to-cell spread. <i>Nature Microbiology</i> , <b>2018</b> , 3, 1472-1485	26.6	40
67	The diaphanous-related formins promote protrusion formation and cell-to-cell spread of Listeria monocytogenes. <i>Journal of Infectious Diseases</i> , <b>2015</b> , 211, 1185-95	7	36
66	Interleukin-6 is associated with steroid resistance and reflects disease activity in severe pediatric ulcerative colitis. <i>Journal of Crohns and Colitis</i> , <b>2013</b> , 7, 916-22	1.5	35
65	Human ALPI deficiency causes inflammatory bowel disease and highlights a key mechanism of gut homeostasis. <i>EMBO Molecular Medicine</i> , <b>2018</b> , 10,	12	33
64	Mutations in Plasmalemma Vesicle Associated Protein Result in Sieving Protein-Losing Enteropathy Characterized by Hypoproteinemia, Hypoalbuminemia, and Hypertriglyceridemia. <i>Cellular and Molecular Gastroenterology and Hepatology</i> , <b>2015</b> , 1, 381-394.e7	7.9	32
63	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> , <b>2020</b> , 158, 2208-2220	13.3	32
62	Mucosa-Associated Ileal Microbiota in New-Onset Pediatric Crohn's Disease. <i>Inflammatory Bowel Diseases</i> , <b>2016</b> , 22, 1533-9	4.5	32
61	Biliary atresia with choledochal cyst: implications for classification. <i>Clinical Gastroenterology and Hepatology</i> , <b>2006</b> , 4, 1411-4	6.9	29
60	TTC7A: Steward of Intestinal Health. <i>Cellular and Molecular Gastroenterology and Hepatology</i> , <b>2019</b> , 7, 555-570	7.9	29
59	Fatal autoimmunity in mice reconstituted with human hematopoietic stem cells encoding defective FOXP3. <i>Blood</i> , <b>2015</b> , 125, 3886-95	2.2	26

58	Large B-Cell Lymphoma in an Adolescent Patient With Interleukin-10 Receptor Deficiency and History of Infantile Inflammatory Bowel Disease. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2016</b> , 63, e15-7	2.8	26
57	Inflammatory Bowel Disease: What Very Early Onset Disease Teaches Us. <i>Gastroenterology Clinics of North America</i> , <b>2018</b> , 47, 755-772	4.4	25
56	Novel de novo mutations of the interleukin-10 receptor gene lead to infantile onset inflammatory bowel disease. <i>Journal of Crohns and Colitis</i> , <b>2014</b> , 8, 1551-6	1.5	24
55	Diagnostic delay in Canadian children with inflammatory bowel disease is more common in Crohn's disease and associated with decreased height. <i>Archives of Disease in Childhood</i> , <b>2018</b> , 103, 319-326	2.2	23
54	Somatic mosaicism and common genetic variation contribute to the risk of very-early-onset inflammatory bowel disease. <i>Nature Communications</i> , <b>2020</b> , 11, 995	17.4	21
53	Anti-TNF, infliximab, and adalimumab can be effective in eosinophilic bowel disease. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2013</b> , 56, 492-7	2.8	21
52	Enzymic characterization of a novel member of the regulatory B-like carboxypeptidase with transcriptional repression function: stimulation of enzymic activity by its target DNA. <i>Biochemical Journal</i> , <b>1999</b> , 343, 341-345	3.8	20
51	Enhanced TH17 Responses in Patients with IL10 Receptor Deficiency and Infantile-onset IBD. <i>Inflammatory Bowel Diseases</i> , <b>2017</b> , 23, 1950-1961	4.5	19
50	Rac2-deficiency leads to exacerbated and protracted colitis in response to <i>Citrobacter rodentium</i> infection. <i>PLoS ONE</i> , <b>2013</b> , 8, e61629	3.7	19
49	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. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2021</b> , 72, 456-473	2.8	19
48	A novel nonsense mutation in the EpCAM gene in a patient with congenital tufting enteropathy. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2014</b> , 58, 18-21	2.8	17
47	Low levels of procalcitonin during episodes of necrotizing enterocolitis. <i>Digestive Diseases and Sciences</i> , <b>2007</b> , 52, 2972-6	4	17
46	CARMIL2 Deficiency Presenting as Very Early Onset Inflammatory Bowel Disease. <i>Inflammatory Bowel Diseases</i> , <b>2019</b> , 25, 1788-1795	4.5	16
45	Multilabel immunofluorescence and antigen reprobing on formalin-fixed paraffin-embedded sections: novel applications for precision pathology diagnosis. <i>Modern Pathology</i> , <b>2016</b> , 29, 557-69	9.8	16
44	Drug Screen Identifies Leflunomide for Treatment of Inflammatory Bowel Disease Caused by TTC7A Deficiency. <i>Gastroenterology</i> , <b>2020</b> , 158, 1000-1015	13.3	16
43	Ankyrin repeat and zinc-finger domain-containing 1 mutations are associated with infantile-onset inflammatory bowel disease. <i>Journal of Biological Chemistry</i> , <b>2017</b> , 292, 7904-7920	5.4	15
42	Replication of genetic variation in the MYO9B gene in Crohn's disease. <i>Human Immunology</i> , <b>2011</b> , 72, 592-7	2.3	15
41	Intensified Infliximab Induction is Associated with Improved Response and Decreased Colectomy in Steroid-Refractory Paediatric Ulcerative Colitis. <i>Journal of Crohns and Colitis</i> , <b>2019</b> , 13, 982-989	1.5	14

40	Long-term outcomes for children with very early-onset colitis: Implications for surgical management. <i>Journal of Pediatric Surgery</i> , <b>2018</b> , 53, 964-967	2.6	14
39	Natural History of Very Early Onset Inflammatory Bowel Disease in North America: A Retrospective Cohort Study. <i>Inflammatory Bowel Diseases</i> , <b>2021</b> , 27, 295-302	4.5	13
38	Very early onset IBD: novel genetic aetiologies. <i>Current Opinion in Allergy and Clinical Immunology</i> , <b>2018</b> , 18, 470-480	3.3	12
37	Mutation spectrum of NOD2 reveals recessive inheritance as a main driver of Early Onset Crohn's Disease. <i>Scientific Reports</i> , <b>2021</b> , 11, 5595	4.9	11
36	Gain-of-function variants in SYK cause immune dysregulation and systemic inflammation in humans and mice. <i>Nature Genetics</i> , <b>2021</b> , 53, 500-510	36.3	11
35	A CARD9 polymorphism is associated with decreased likelihood of persistent conjugated hyperbilirubinemia in intestinal failure. <i>PLoS ONE</i> , <b>2014</b> , 9, e85915	3.7	9
34	Monogenic Intestinal Epithelium Defects and the Development of Inflammatory Bowel Disease. <i>Physiology</i> , <b>2018</b> , 33, 360-369	9.8	8
33	Rac1 Polymorphisms and Thiopurine Efficacy in Children With Inflammatory Bowel Disease. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2015</b> , 61, 404-7	2.8	8
32	Unrelated donor hematopoietic stem cell transplantation for infantile enteropathy due to IL-10/IL-10 receptor defect. <i>Pediatric Transplantation</i> , <b>2015</b> , 19, E101-3	1.8	7
31	Association between a multi-locus genetic risk score and inflammatory bowel disease. <i>Bioinformatics and Biology Insights</i> , <b>2013</b> , 7, 143-52	5.3	7
30	Apical junction complex proteins and ulcerative colitis: a focus on the PTPRS gene. <i>Expert Review of Molecular Diagnostics</i> , <b>2008</b> , 8, 465-77	3.8	7
29	Diagnostic Delay Is Associated With Complicated Disease and Growth Impairment in Paediatric Crohn's Disease. <i>Journal of Crohns and Colitis</i> , <b>2021</b> , 15, 419-431	1.5	6
28	The risk of myocardial infarction in HIV-infected patients receiving HAART: a case report. <i>International Journal of STD and AIDS</i> , <b>2001</b> , 12, 612-3	1.4	5
27	A Systematic Review of Monogenic Inflammatory Bowel Disease. <i>Clinical Gastroenterology and Hepatology</i> , <b>2021</b> ,	6.9	5
26	Novel CARMIL2 loss-of-function variants are associated with pediatric inflammatory bowel disease. <i>Scientific Reports</i> , <b>2021</b> , 11, 5945	4.9	5
25	NOX1 Regulates Collective and Planktonic Cell Migration: Insights From Patients With Pediatric-Onset IBD and NOX1 Deficiency. <i>Inflammatory Bowel Diseases</i> , <b>2020</b> , 26, 1166-1176	4.5	4
24	An integrated taxonomy for monogenic inflammatory bowel disease. <i>Gastroenterology</i> , <b>2021</b> ,	13.3	4
23	Novel Exonic Deletions in TTC7A in a Newborn with Multiple Intestinal Atresia and Combined Immunodeficiency. <i>Journal of Clinical Immunology</i> , <b>2019</b> , 39, 616-619	5.7	3

22	Human autoinflammatory disease reveals ELF4 as a transcriptional regulator of inflammation. <i>Nature Immunology</i> , <b>2021</b> , 22, 1118-1126	19.1	3
21	Cutting Edge: NOX2 NADPH Oxidase Controls Infection by an Intracellular Bacterial Pathogen through Limiting the Type 1 IFN Response. <i>Journal of Immunology</i> , <b>2021</b> , 206, 323-328	5.3	3
20	Predictive Prenatal Diagnosis for Infantile-onset Inflammatory Bowel Disease Because of Interleukin-10 Signalling Defects. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2021</b> , 72, 276-281	2.8	3
19	Advanced Understanding of Monogenic Inflammatory Bowel Disease. <i>Frontiers in Pediatrics</i> , <b>2020</b> , 8, 618918	3.4	3
18	Deficiency in X-linked inhibitor of apoptosis protein promotes susceptibility to microbial triggers of intestinal inflammation. <i>Science Immunology</i> , <b>2021</b> , 6, eabf7473	2.8	2
17	Mutation spectrum of NOD2 reveals recessive inheritance as a main driver of Early Onset Crohn's Disease		2
16	The E3 ubiquitin ligase UBR5 interacts with TTC7A and may be associated with very early onset inflammatory bowel disease. <i>Scientific Reports</i> , <b>2020</b> , 10, 18648	4.9	2
15	ARPC1B binds WASP to control actin polymerization and curtail tonic signaling in B cells. <i>JCI Insight</i> , <b>2021</b> , 6,	9.9	1
14	Multisystem Autoimmune Inflammatory Disease, Including Colitis, Due to Inborn Error of Immunity. <i>Pediatrics</i> , <b>2021</b> , 148,	7.4	1
13	Utilization of Whole Exome Sequencing Data to Identify Clinically Relevant Pharmacogenomic Variants in Pediatric Inflammatory Bowel Disease. <i>Clinical and Translational Gastroenterology</i> , <b>2020</b> , 11, e00263	4.2	1
12	Application of Whole Exome Sequencing in Congenital Secretory Diarrhea Diagnosis. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2019</b> , 68, e106-e108	2.8	1
11	Whipple disease mimicking inflammatory bowel disease. <i>Intestinal Research</i> , <b>2021</b> , 19, 119-125	4.1	1
10	Variants in STXBP3 are Associated with Very Early Onset Inflammatory Bowel Disease, Bilateral Sensorineural Hearing Loss and Immune Dysregulation. <i>Journal of Crohn's and Colitis</i> , <b>2021</b> , 15, 1908-1919	1.5	0
9	Very Early Onset Inflammatory Bowel Disease (VEOIBD) <b>2019</b> , 383-404		
8	2019 Harry Shwachman Award: Dr Anne M. Griffiths, MD, FRCPC. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2020</b> , 70, 405	2.8	
7	Inflammatory Bowel Disease in Primary Immunodeficiencies <b>2018</b> , 167-181		
6	NADPH oxidase complex and IBD Candidate Gene studies O-18.. <i>Inflammatory Bowel Diseases</i> , <b>2011</b> , 17, S8	4.5	
5	The Authors' reply: Figure 1. <i>Gut</i> , <b>2012</b> , 61, 1097.2-1098	19.2	

4 A Machine Learning Approach to Identifying Causal Monogenic Variants in Inflammatory Bowel Disease **2022**, 1, 171-179

3 Sequencing and Mapping IBD Genes to Individual Causative Variants and Their Clinical Relevance **2019**, 117-139

2 Pediatric Diarrheal Disorders **2020**, 143-157

1 Valosin-containing protein-regulated endoplasmic reticulum stress causes NOD2-dependent inflammatory responses.. *Scientific Reports*, **2022**, 12, 3906

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