Siobhan O Burns

List of Publications by Year
in descending order

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112 papers	7,360 citations	61984 43 h-index	60623 81 g-index
118 all docs	118 docs citations	118 times ranked	10790 citing authors

#	Article	IF	Citations
1	A robust model for read count data in exome sequencing experiments and implications for copy number variant calling. Bioinformatics, 2012, 28, 2747-2754.	4.1	534
2	Clinical spectrum and features of activated phosphoinositide 3-kinase Î´ syndrome: AÂlarge patient cohort study. Journal of Allergy and Clinical Immunology, 2017, 139, 597-606.e4.	2.9	377
3	WASP: a key immunological multitasker. Nature Reviews Immunology, 2010, 10, 182-192.	22.7	354
4	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	6.2	343
5	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
6	Configuration of human dendritic cell cytoskeleton by Rho GTPases, the WAS protein, and differentiation. Blood, 2001, 98, 1142-1149.	1.4	300
7	The extended phenotype of LPS-responsive beige-like anchor protein (LRBA) deficiency. Journal of Allergy and Clinical Immunology, 2016, 137, 223-230.	2.9	247
8	Two novel activating mutations in the Wiskott-Aldrich syndrome protein result in congenital neutropenia. Blood, 2006, 108, 2182-2189.	1.4	200
9	COVID-19 in patients with primary and secondary immunodeficiency: The United Kingdom experience. Journal of Allergy and Clinical Immunology, 2021, 147, 870-875.e1.	2.9	188
10	Loss-of-function nuclear factor ÎºB subunit 1 (NFKB1) variants are the most common monogenic cause of common variable immunodeficiency in Europeans. Journal of Allergy and Clinical Immunology, 2018, 142, 1285-1296.	2.9	185
11	X-linked thrombocytopenia (XLT) due to WAS mutations: clinical characteristics, long-term outcome, and treatment options. Blood, 2010, 115, 3231-3238.	1.4	178
12	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. Nature, 2020, 583, 90-95.	27.8	148
13	British Lung Foundation/United Kingdom Primary Immunodeficiency Network Consensus Statement on the Definition, Diagnosis, and Management of Granulomatous-Lymphocytic Interstitial Lung Disease in Common Variable Immunodeficiency Disorders. Journal of Allergy and Clinical Immunology: in Practice, 2017, 5, 938-945.	3.8	138
14	Maturation of DC is associated with changes in motile characteristics and adherence. Cytoskeleton, 2004, 57, 118-132.	4.4	137
15	Disease Evolution and Response to Rapamycin in Activated Phosphoinositide 3-Kinase Î´ Syndrome: The European Society for Immunodeficiencies-Activated Phosphoinositide 3-Kinase Î´ Syndrome Registry. Frontiers in Immunology, 2018, 9, 543.	4.8	137
16	The leukocyte podosome. European Journal of Cell Biology, 2006, 85, 151-157.	3.6	135
17	Mechanisms of WASp-mediated hematologic and immunologic disease. Blood, 2004, 104, 3454-3462.	1.4	134
18	Immune deficiency and autoimmunity in patients with CTLA-4 (CD152) mutations. Clinical and Experimental Immunology, 2017, 190, 1-7.	2.6	123

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19	Clinical and Immunological Phenotype of Patients With Primary Immunodeficiency Due to Damaging Mutations in NFKB2. Frontiers in Immunology, 2019, 10, 297.	4.8	117
20	WIP Regulates the Stability and Localization of WASP to Podosomes in Migrating Dendritic Cells. Current Biology, 2006, 16, 2337-2344.	3.9	114
21	Biallelic JAK1 mutations in immunodeficient patient with mycobacterial infection. Nature Communications, 2016, 7, 13992.	12.8	104
22	Identifying functional defects in patients with immune dysregulation due to LRBA and CTLA-4 mutations. Blood, 2017, 129, 1458-1468.	1.4	102
23	B cellâ€“intrinsic deficiency of the Wiskott-Aldrich syndrome protein (WASp) causes severe abnormalities of the peripheral B-cell compartment in mice. Blood, 2012, 119, 2819-2828.	1.4	99
24	Inherited p40phox deficiency differs from classic chronic granulomatous disease. Journal of Clinical Investigation, 2018, 128, 3957-3975.	8.2	99
25	Wiskottâ€“Aldrich Syndrome: Immunodeficiency resulting from defective cell migration and impaired immunostimulatory activation. Immunobiology, 2009, 214, 778-790.	1.9	90
26	LRBA gene deletion in a patient presenting with autoimmunity without hypogammaglobulinemia. Journal of Allergy and Clinical Immunology, 2012, 130, 1428-1432.	2.9	90
27	Impaired T-cell priming in vivo resulting from dysfunction of WASp-deficient dendritic cells. Blood, 2007, 110, 4278-4284.	1.4	86
28	Gene therapy for Wiskott-Aldrich syndrome in a severely affected adult. Blood, 2017, 130, 1327-1335.	1.4	83
29	Activating PI3KÎ´ mutations in a cohort of 669 patients with primary immunodeficiency. Clinical and Experimental Immunology, 2016, 183, 221-229.	2.6	82
30	Characterization of the clinical and immunologic phenotype and management of 157 individuals with 56 distinct heterozygous NFKB1 mutations. Journal of Allergy and Clinical Immunology, 2020, 146, 901-911.	2.9	78
31	A new functional assay for the diagnosis of X-linked inhibitor of apoptosis (XIAP) deficiency. Clinical and Experimental Immunology, 2014, 176, 394-400.	2.6	75
32	Cytoskeletal remodeling mediated by WASp in dendritic cells is necessary for normal immune synapse formation and T-cell priming. Blood, 2011, 118, 2492-2501.	1.4	73
33	Successful outcome following allogeneic hematopoietic stem cell transplantation in adults with primary immunodeficiency. Blood, 2018, 131, 917-931.	1.4	68
34	Immunodeficiency and severe susceptibility to bacterial infection associated with a loss-of-function homozygous mutation of MKL1. Blood, 2015, 126, 1527-1535.	1.4	66
35	Extended clinical and immunological phenotype and transplant outcome in CD27 and CD70 deficiency. Blood, 2020, 136, 2638-2655.	1.4	64
36	Phosphorylation of WASp is a key regulator of activity and stability in vivo. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 15738-15743.	7.1	51

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37	Wiskott-Aldrich syndrome protein regulates autophagy and inflammasome activity in innate immune cells. Nature Communications, 2017, 8, 1576.	12.8	50
38	Recent advances in the understanding of genetic defects of neutrophil number and function. British Journal of Haematology, 2010, 151, 312-326.	2.5	49
39	Transmission of Hepatitis B Core Antibody and Galactomannan Enzyme Immunoassay Positivity via Immunoglobulin Products: A Comprehensive Analysis. Clinical Infectious Diseases, 2016, 63, 57-63.	5.8	49
40	Inflammatory and autoimmune manifestations in X-linked carriers of chronic granulomatous disease in the United Kingdom. Journal of Allergy and Clinical Immunology, 2017, 140, 628-630.e6.	2.9	48
41	Respiratory Infections and Antibiotic Usage in Common Variable Immunodeficiency. Journal of Allergy and Clinical Immunology: in Practice, 2018, 6, 159-168.e3.	3.8	46
42	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. American Journal of Human Genetics, 2018, 103, 3-18.	6.2	46
43	The Wiskott-Aldrich syndrome: disordered actin dynamics in haematopoietic cells. Immunological Reviews, 2000, 178, 118-128.	6.0	45
44	Clinical and laboratory features of seventy-eight UK patients with Goodâ€™s syndrome (thymoma and) Tj ETQq0 0 0 rgBT /Overlock 10	2.6	45
45	A Partial Down-regulation of WASP Is Sufficient to Inhibit Podosome Formation in Dendritic Cells. Molecular Therapy, 2006, 13, 729-737.	8.2	44
46	Mutagenesis in Norovirus in Response to Favipiravir Treatment. New England Journal of Medicine, 2018, 379, 2173-2176.	27.0	43
47	A case of XMEN syndrome presented with severe auto-immune disorders mimicking autoimmune lymphoproliferative disease. Clinical Immunology, 2015, 159, 58-62.	3.2	41
48	Natural Killer Cells from Patients with Recombinase-Activating Gene and Non-Homologous End Joining Gene Defects Comprise a Higher Frequency of CD56bright NKG2A+++ Cells, and Yet Display Increased Degranulation and Higher Perforin Content. Frontiers in Immunology, 2017, 8, 798.	4.8	41
49	Prevalence and clinical challenges among adults with primary immunodeficiency and recombination-activating gene deficiency. Journal of Allergy and Clinical Immunology, 2018, 141, 2303-2306.	2.9	40
50	Recombination activity of human recombination-activating gene 2 (RAG2) mutations and correlation with clinical phenotype. Journal of Allergy and Clinical Immunology, 2019, 143, 726-735.	2.9	39
51	Immunodeficiency and disseminated mycobacterial infection associated with homozygous nonsense mutation of IKKÎ². Journal of Allergy and Clinical Immunology, 2014, 134, 215-218.e3.	2.9	37
52	How I manage patients with Wiskott Aldrich syndrome. British Journal of Haematology, 2019, 185, 647-655.	2.5	37
53	SARS-CoV-2 Vaccine Responses in Individuals with Antibody Deficiency: Findings from the COV-AD Study. Journal of Clinical Immunology, 2022, 42, 923-934.	3.8	37
54	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36

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55	Impaired neutrophil migration and phagocytosis in IRAKâ€4 deficiency. British Journal of Haematology, 2009, 147, 153-156.	2.5	32
56	Deletion of Wiskottâ€“Aldrich syndrome protein triggers Rac2 activity and increased cross-presentation by dendritic cells. Nature Communications, 2016, 7, 12175.	12.8	31
57	Atypical Severe Combined Immunodeficiency Caused by a Novel Homozygous Mutation In Rag1 Gene in a Girl who Presented with Pyoderma Gangrenosum: A Case Report and Literature Review. Journal of Clinical Immunology, 2014, 34, 792-795.	3.8	30
58	Study of an extended family with CTLA-4 deficiency suggests a CD28/CTLA-4 independent mechanism responsible for differences in disease manifestations and severity. Clinical Immunology, 2018, 188, 94-102.	3.2	30
59	Bronchiectasis and deteriorating lung function in agammaglobulinaemia despite immunoglobulin replacement therapy. Clinical and Experimental Immunology, 2018, 191, 212-219.	2.6	30
60	Primary immunodeficiencies due to abnormalities of the actin cytoskeleton. Current Opinion in Hematology, 2017, 24, 16-22.	2.5	29
61	Modelling human wiskott aldrich syndrome protein mutants in zebrafish larvae using live in vivo imaging. Journal of Cell Science, 2013, 126, 4077-84.	2.0	28
62	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	6.2	26
63	X-linked Inhibitor of Apoptosis Complicated by Granulomatous Lymphocytic Interstitial Lung Disease (GLILD) and Granulomatous Hepatitis. Journal of Clinical Immunology, 2016, 36, 733-738.	3.8	25
64	Outcomes following SARS-CoV-2 infection in patients with primary and secondary immunodeficiency in the UK. Clinical and Experimental Immunology, 2022, 209, 247-258.	2.6	25
65	A comprehensive characterization of chronic norovirus infection in immunodeficient hosts. Journal of Allergy and Clinical Immunology, 2019, 144, 1450-1453.	2.9	24
66	Key diagnostic markers for autoimmune lymphoproliferative syndrome with molecular genetic diagnosis. Blood, 2020, 136, 1933-1945.	1.4	24
67	Dendritic Cells: The Bare Bones of Immunity. Current Biology, 2004, 14, R965-R967.	3.9	22
68	Exacerbated experimental arthritis in Wiskottâ€“Aldrich syndrome protein deficiency: Modulatory role of regulatory B cells. European Journal of Immunology, 2014, 44, 2692-2702.	2.9	22
69	Lentiviral Vector-Mediated Correction of a Mouse Model of Leukocyte Adhesion Deficiency Type I. Human Gene Therapy, 2016, 27, 668-678.	2.7	21
70	B cellâ€“intrinsic requirement for STK4 in humoral immunity in mice and human subjects. Journal of Allergy and Clinical Immunology, 2019, 143, 2302-2305.	2.9	21
71	Expanding Clinical Phenotype and Novel Insights into the Pathogenesis of ICOS Deficiency. Journal of Clinical Immunology, 2020, 40, 277-288.	3.8	21
72	Paediatric hereditary angioedema: a survey of UK service provision and patient experience. Clinical and Experimental Immunology, 2014, 178, 483-488.	2.6	19

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73	Anti-MÃ¼llerian hormone and Inhibin B after stem cell transplant in childhood: a comparison of myeloablative, reduced intensity and treosulfan-based chemotherapy regimens. Bone Marrow Transplantation, 2020, 55, 1985-1995.	2.4	19
74	Topoisomerase 2Î² mutation impairs early B-cell development. Blood, 2020, 135, 1497-1501.	1.4	18
75	Altered Microbiota, Impaired Quality of Life, Malabsorption, Infection, and Inflammation in CVID Patients With Diarrhoea. Frontiers in Immunology, 2020, 11, 1654.	4.8	17
76	Curation and expansion of Human Phenotype Ontology for defined groups of inborn errors of immunity. Journal of Allergy and Clinical Immunology, 2022, 149, 369-378.	2.9	16
77	Low seropositivity and suboptimal neutralisation rates in patients fully vaccinated against COVIDâ€19 with Bâ€cell malignancies. British Journal of Haematology, 2021, 195, 706-709.	2.5	16
78	How I use allogeneic HSCT for adults with inborn errors of immunity. Blood, 2021, 138, 1666-1676.	1.4	15
79	Phenotype, genotype, treatment, and survival outcomes in patients with X-linked inhibitor of apoptosis deficiency. Journal of Allergy and Clinical Immunology, 2022, 150, 456-466.	2.9	15
80	A congenital activating mutant of WASp causes altered plasma membrane topography and adhesion under flow in lymphocytes. Blood, 2010, 115, 5355-5365.	1.4	14
81	Genetic Variants Associated With Neutrophil Function in Aggressive Periodontitis and Healthy Controls. Journal of Periodontology, 2010, 81, 527-534.	3.4	14
82	Common variable immunodeficiency and natural killer cell lymphopenia caused by Ets-binding site mutation in the IL-2 receptor Î³ (IL2RG) gene promoter. Journal of Allergy and Clinical Immunology, 2016, 137, 940-942.e4.	2.9	14
83	An intronic deletion in megakaryoblastic leukemia 1 is associated with hyperproliferation of B cells in triplets with Hodgkin lymphoma. Haematologica, 2020, 105, 1339-1350.	3.5	13
84	Absence of Î³-Chain in Keratinocytes Alters Chemokine Secretion, Resulting in Reduced Immune Cell Recruitment. Journal of Investigative Dermatology, 2017, 137, 2120-2130.	0.7	12
85	Is It Safe to Switch From Intravenous Immunoglobulin to Subcutaneous Immunoglobulin in Patients With Common Variable Immunodeficiency and Autoimmune Thrombocytopenia?. Frontiers in Immunology, 2018, 9, 1656.	4.8	12
86	Clinical Phenotype and Long Term Outcome in a Large Cohort of X-Linked Thrombocytopenia (XLT)/Mild Wiskott-Aldrich-Syndrome Patients. Blood, 2008, 112, 90-90.	1.4	12
87	Disease-associated missense mutations in the EVH1 domain disrupt intrinsic WASp function causing dysregulated actin dynamics and impaired dendritic cell migration. Blood, 2013, 121, 72-84.	1.4	11
88	Lentiviral gene therapy rescues p47phox chronic granulomatous disease and the ability to fight Salmonella infection in mice. Gene Therapy, 2020, 27, 459-469.	4.5	11
89	Activating mutations of N-WASP alter Shigella pathogenesis. Biochemical and Biophysical Research Communications, 2009, 384, 284-289.	2.1	10
90	Bleeding and splenectomy in Wiskott-Aldrich syndrome: A single-centre experience. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 1042-1044.e1.	3.8	10

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91	Managing Granulomatousâ€“Lymphocytic Interstitial Lung Disease in Common Variable Immunodeficiency Disorders: e-GLILDnet International Clinicians Survey. Frontiers in Immunology, 2020, 11, 606333.	4.8	10
92	Long-term outcomes for adults with chronic granulomatous disease in the United Kingdom. Journal of Allergy and Clinical Immunology, 2021, 147, 1104-1107.	2.9	10
93	Bowel Histology of CVID Patients Reveals Distinct Patterns of Mucosal Inflammation. Journal of Clinical Immunology, 2022, 42, 46-59.	3.8	10
94	Retrospective, Landmark Analysis of Long-term Adult Morbidity Following Allogeneic HSCT for Inborn Errors of Immunity in Infancy and Childhood. Journal of Clinical Immunology, 2022, 42, 1230-1243.	3.8	10
95	Loss of Janus Associated Kinase 1 Alters Urothelial Cell Function and Facilitates the Development of Bladder Cancer. Frontiers in Immunology, 2019, 10, 2065.	4.8	9
96	Health-Related Quality of Life and Emotional Health in X-Linked Carriers of Chronic Granulomatous Disease in the United Kingdom. Journal of Clinical Immunology, 2019, 39, 195-199.	3.8	9
97	Airway inflammation and dysbiosis in antibody deficiency despite the presence of IgG. Journal of Allergy and Clinical Immunology, 2022, 149, 2105-2115.e10.	2.9	8
98	Wiskott-Aldrich syndrome: a disorder of haematopoietic cytoskeletal regulation. , 1999, 47, 107-113.		7
99	Dendritic cell-expressed common gamma-chain recruits IL-15 for trans-presentation at the murine immunological synapse. Wellcome Open Research, 2018, 3, 84.	1.8	7
100	FAS mutations are an uncommon cause of immune thrombocytopenia in children and adults without additional features of immunodeficiency. British Journal of Haematology, 2019, 186, e163-e165.	2.5	6
101	Granulomatousâ€“lymphocytic interstitial lung disease: an international research prioritisation. ERJ Open Research, 2021, 7, 00467-2021.	2.6	6
102	Lossâ€ofâ€function mutations in CSF3R cause moderate neutropenia with fully mature neutrophils: two novel pedigrees. British Journal of Haematology, 2020, 191, 930-934.	2.5	5
103	Overactive WASp in X-linked neutropenia leads to aberrant B-cell division and accelerated plasma cell generation. Journal of Allergy and Clinical Immunology, 2022, 149, 1069-1084.	2.9	5
104	Predictive Factors for and Complications of Bronchiectasis in Common Variable Immunodeficiency Disorders. Journal of Clinical Immunology, 2022, 42, 572-581.	3.8	5
105	Dendritic cell-expressed common gamma-chain recruits IL-15 for trans-presentation at the murine immunological synapse. Wellcome Open Research, 2018, 3, 84.	1.8	4
106	Lentivectors are efficient tools to manipulate the dendritic cell cytoskeleton. Cytoskeleton, 2011, 68, 434-445.	2.0	3
107	Dendritic cell defects in primary immunodeficiency disorders. LymphoSign Journal, 2016, 3, 1-12.	0.2	2
108	Neutrophil dysfunction in children. Paediatrics and Child Health (United Kingdom), 2010, 20, 531-538.	0.4	1

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109	Transverse myelitis in a patient with activated phosphoinositide 3-kinase Î´ syndrome type 1. Clinical Immunology, 2020, 219, 108552.	3.2	1
110	A Case of Burkholderia Prostatitis in a Patient with Chronic Granulomatous Disease. Journal of Clinical Immunology, 2020, 40, 1204-1206.	3.8	1
111	P29â€ƒUse of novel biologic therapies for arthropathy affecting common variable immunodeficiency patients. Rheumatology, 2020, 59, .	1.9	0
112	Malignancies in Cellular Immunodeficiencies. Rare Diseases of the Immune System, 2021, , 361-389.	0.1	0