Roshini S Abraham

List of Publications by Year
in descending order

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115 papers	4,882 citations	147801 31 h-index	102487 66 g-index
118 all docs	118 docs citations	118 times ranked	6607 citing authors

#	Article	IF	Citations
1	Supporting Careers of Women in Clinical Immunology: From Conceptualization to Implementation. Frontiers in Pediatrics, 2022, 10, 864734.	1.9	1
2	Interpretation of Dihydrorhodamine-1,2,3 Flow Cytometry in Chronic Granulomatous Disease: an Atypical Exemplar. Journal of Clinical Immunology, 2022, , 1.	3.8	0
3	Defining the mild variant of leukocyte adhesion deficiency type <scp>II</scp> (<scp>SLC35C1</scp>â€congenital disorder of glycosylation) and response to <scp>l</scp>â€fucose therapy: Insights from two new families and review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 2005-2018.	1.2	10
4	Case report and review of the literature: immune dysregulation in a large familial cohort due to a novel pathogenicRELAvariant. Rheumatology, 2022, 62, 347-359.	1.9	4
5	POLD1 Deficiency Reveals a Role for POLD1 in DNA Repair and T and B Cell Development. Journal of Clinical Immunology, 2021, 41, 270-273.	3.8	10
6	Severe SARS-CoV-2 disease in the context of a NF-Î¹B2 loss-of-function pathogenic variant. Journal of Allergy and Clinical Immunology, 2021, 147, 532-544.e1.	2.9	25
7	Coronavirus disease 2019 in patients with inborn errors of immunity: An international study. Journal of Allergy and Clinical Immunology, 2021, 147, 520-531.	2.9	278
8	Functional Confirmation of DNA Repair Defect in Ataxia Telangiectasia (AT) Infants Identified by Newborn Screening for Severe Combined Immunodeficiency (NBS SCID). Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 723-732.e3.	3.8	8
9	The New â€œWholly Trinityâ€•in the Diagnosis and Management of Inborn Errors of Immunity. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 613-625.	3.8	10
10	Infectious Complications Predict Premature CD8+ T-cell Senescence in CD40 Ligand-Deficient Patients. Journal of Clinical Immunology, 2021, 41, 795-806.	3.8	2
11	Immune Reconstitution after Hematopoietic Stem Cell Transplantation in Immunodeficiencyâ€“Centromeric Instabilityâ€“Facial Anomalies Syndrome Type 1. Journal of Clinical Immunology, 2021, 41, 1089-1094.	3.8	2
12	Next-generation sequencing for inborn errors of immunity. Human Immunology, 2021, 82, 871-882.	2.4	12
13	Delayed-Onset ADA1 (ADA) Deficiency Not Detected by TREC Screen. Pediatrics, 2021, 147, e2020005579.	2.1	3
14	Pneumococcal serotype-specific cut-offs based on antibody responses to pneumococcal polysaccharide vaccination in healthy adults. Vaccine, 2021, 39, 2850-2856.	3.8	7
15	Do monogenic inborn errors of immunity cause susceptibility to severe COVID-19?. Journal of Clinical Investigation, 2021, 131, .	8.2	3
16	Identification of 22 novel BTK gene variants in B cell deficiency with hypogammaglobulinemia. Clinical Immunology, 2021, 229, 108788.	3.2	2
17	Sex differences in antibody responses to the 23-valent pneumococcal polysaccharide vaccination. Annals of Allergy, Asthma and Immunology, 2021, 127, 509-510.	1.0	3
18	Rubella Virus Infected Macrophages and Neutrophils Define Patterns of Granulomatous Inflammation in Inborn and Acquired Errors of Immunity. Frontiers in Immunology, 2021, 12, 796065.	4.8	19

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19	Relationship between asthma status and antibody response pattern to 23-valent pneumococcal vaccination. Journal of Asthma, 2020, 57, 381-390.	1.7	8
20	NAPDH Oxidase-Specific Flow Cytometry Allows for Rapid Genetic Triage and Classification of Novel Variants in Chronic Granulomatous Disease. Journal of Clinical Immunology, 2020, 40, 191-202.	3.8	8
21	How to evaluate for immunodeficiency in patients with autoimmune cytopenias: laboratory evaluation for the diagnosis of inborn errors of immunity associated with immune dysregulation. Hematology American Society of Hematology Education Program, 2020, 2020, 661-672.	2.5	10
22	Follow-Up for an Abnormal Newborn Screen for Severe Combined Immunodeficiencies (NBS SCID): A Clinical Immunology Society (CIS) Survey of Current Practices. International Journal of Neonatal Screening, 2020, 6, 52.	3.2	12
23	A Novel Pathogenic Variant in CARMIL2 (RLTPR) Causing CARMIL2 Deficiency and EBV-Associated Smooth Muscle Tumors. Frontiers in Immunology, 2020, 11, 884.	4.8	26
24	PAX1 is essential for development and function of the human thymus. Science Immunology, 2020, 5, .	11.9	55
25	Editorial: Application of Cytometry in Primary Immunodeficiencies. Frontiers in Immunology, 2020, 11, 463.	4.8	4
26	Cryptococcal pneumonia in an adolescent with a gain-of-function variant in signal transduction and activator of transcription 1 (STAT1). BMJ Case Reports, 2020, 13, e234120.	0.5	3
27	Expanding mechanistic insights into the pathogenesis of idiopathic CD4+ T cell lymphocytopenia. Journal of Clinical Investigation, 2020, 130, 5105-5108.	8.2	1
28	Clinical Correlation of Cytomegalovirus Infection With CMV-specific CD8+ T-cell Immune Competence Score and Lymphocyte Subsets in Solid Organ Transplant Recipients. Transplantation, 2019, 103, 832-838.	1.0	29
29	â€œThe State of the Unionâ€: Current and Future Perspectives on Patient-Centric Care for Primary Immunodeficiencies and Immune Dysregulatory Diseases. Frontiers in Immunology, 2019, 10, 1783.	4.8	2
30	Phenotypic heterogeneity associated with germline GATA2 haploinsufficiency: a comprehensive kindred study. Leukemia and Lymphoma, 2019, 60, 3282-3286.	1.3	4
31	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. American Journal of Human Genetics, 2019, 105, 108-121.	6.2	39
32	Provider Perceptions of Quality of Life, Neurocognition, Physical Well-being, and Psychosocial Health in Patients with Primary Immunodeficiency/Immune Dysregulation Conditions. Journal of Clinical Immunology, 2019, 39, 805-813.	3.8	3
33	Outcomes and Treatment Strategies for Autoimmunity and Hyperinflammation in Patients with RAG Deficiency. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 1970-1985.e4.	3.8	64
34	A Systematic Review on Predisposition to Lymphoid (B and T cell) Neoplasias in Patients With Primary Immunodeficiencies and Immune Dysregulatory Disorders (Inborn Errors of Immunity). Frontiers in Immunology, 2019, 10, 777.	4.8	59
35	X-Linked Lymphoproliferative Syndrome Presenting as Adult-Onset Multi-Infarct Dementia. Journal of Neuropathology and Experimental Neurology, 2019, 78, 460-466.	1.7	6
36	Activating mutations in PIK3CD disrupt the differentiation and function of human and murine CD4+ T cells. Journal of Allergy and Clinical Immunology, 2019, 144, 236-253.	2.9	44

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37	The total IgM, IgA and IgG antibody responses to pneumococcal polysaccharide vaccination (PneumovaxÂ®23) in a healthy adult population and patients diagnosed with primary immunodeficiencies. Vaccine, 2019, 37, 1350-1355.	3.8	15
38	Diagnostic Tools for Inborn Errors of Human Immunity (Primary Immunodeficiencies and Immune) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	5.3	15
39	Use of Genetic Testing for Primary Immunodeficiency Patients. Journal of Clinical Immunology, 2018, 38, 320-329.	3.8	88
40	Utility of DNA, RNA, Protein, and Functional Approaches to Solve Cryptic Immunodeficiencies. Journal of Clinical Immunology, 2018, 38, 307-319.	3.8	29
41	Consequences of B-cell-depleting therapy: hypogammaglobulinemia and impaired B-cell reconstitution. Immunotherapy, 2018, 10, 713-728.	2.0	93
42	Clinical spectrum and clonal evolution in germline syndromes with predisposition to myeloid neoplasms. British Journal of Haematology, 2018, 182, 141-145.	2.5	4
43	Successful Nonmyeloablative Allogeneic Stem Cell Transplant in a Child With Emberger Syndrome and GATA2 Mutation. Journal of Pediatric Hematology/Oncology, 2018, 40, e383-e388.	0.6	10
44	Optimal approach to assessing T-cell function in haematopoietic cell transplant recipients. BMJ Case Reports, 2018, 2018, bcr-2017-222417.	0.5	2
45	Clinical Correlates and Treatment Outcomes for Patients With Short Telomere Syndromes. Mayo Clinic Proceedings, 2018, 93, 834-839.	3.0	20
46	Diagnostic Utility of Complement Serology for Atypical Hemolytic Uremic Syndrome. Mayo Clinic Proceedings, 2018, 93, 1351-1362.	3.0	17
47	Application of a radiosensitivity flow assay in a patient with DNA ligase 4 deficiency. Blood Advances, 2018, 2, 1828-1832.	5.2	13
48	Defective TLR9-driven STAT3 activation in B cells of patients with CVID. Clinical Immunology, 2018, 197, 40-44.	3.2	7
49	Short telomere syndromes cause a primary T cell immunodeficiency. Journal of Clinical Investigation, 2018, 128, 5222-5234.	8.2	82
50	Gastrointestinal Manifestations in X-linked Agammaglobulinemia. Journal of Clinical Immunology, 2017, 37, 287-294.	3.8	51
51	A case of atypical, complete DiGeorge syndrome without 22q11Âmutation. Annals of Allergy, Asthma and Immunology, 2017, 118, 640-642.e2.	1.0	6
52	WILD syndrome is GATA2 deficiency: A novel deletion in the GATA2 gene. Journal of Allergy and Clinical Immunology: in Practice, 2017, 5, 1149-1152.e1.	3.8	16
53	A Droplet Digital PCR Method for Severe Combined Immunodeficiency Newborn Screening. Journal of Molecular Diagnostics, 2017, 19, 755-765.	2.8	14
54	Unrelated Hematopoietic Cell Transplantation in a Patient with Combined Immunodeficiency with Granulomatous Disease and Autoimmunity Secondary to RAG Deficiency. Journal of Clinical Immunology, 2016, 36, 725-732.	3.8	19

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55	Thrombotic Microangiopathy Care Pathway: A Consensus Statement for the Mayo Clinic Complement Alternative Pathway-Thrombotic Microangiopathy (CAP-TMA) Disease-Oriented Group. Mayo Clinic Proceedings, 2016, 91, 1189-1211.	3.0	55
56	RNA sequencing reveals the consequences of a novel insertion in dedicator of cytokinesis-8. Journal of Allergy and Clinical Immunology, 2016, 138, 289-292.e6.	2.9	6
57	Flow Cytometry, a Versatile Tool for Diagnosis and Monitoring of Primary Immunodeficiencies. Vaccine Journal, 2016, 23, 254-271.	3.1	76
58	Outcome of Whole Exome Sequencing for Diagnostic Odyssey Cases of an Individualized Medicine Clinic. Mayo Clinic Proceedings, 2016, 91, 297-307.	3.0	83
59	Diagnostic accuracy and clinical relevance of an inflammatory biomarker panel for sepsis in adult critically ill patients. Diagnostic Microbiology and Infectious Disease, 2016, 84, 175-180.	1.8	25
60	Spectrum of myeloid neoplasms and immune deficiency associated with germline <scp>GATA</scp>2 mutations. Cancer Medicine, 2015, 4, 490-499.	2.8	43
61	Combinatorial Immunoprofiling in Latent Tuberculosis Infection. Toward Better Risk Stratification. American Journal of Respiratory and Critical Care Medicine, 2015, 192, 605-617.	5.6	28
62	Newborn Screening for Severe Combined Immunodeficiency: Changing the Landscape of Pediatric Primary Immunodeficiencies. Journal of Allergy and Clinical Immunology: in Practice, 2015, 3, 1008-1009.	3.8	1
63	Monocyte HLA-DR expression and neutrophil CD64 expression as biomarkers of infection in critically ill neonates and infants. Pediatric Research, 2015, 78, 683-690.	2.3	23
64	Dermatologic Features of ADA2 Deficiency in Cutaneous Polyarteritis Nodosa. JAMA Dermatology, 2015, 151, 1230.	4.1	75
65	Broad-spectrum antibodies against self-antigens and cytokines in RAG deficiency. Journal of Clinical Investigation, 2015, 125, 4135-4148.	8.2	159
66	Analysis of Serum Ferritin Levels As a Diagnostic Criteria for Hemophagocytic Lymphohistiocytosis (HLH) in Hospitalized Adult Patients. Blood, 2015, 126, 1014-1014.	1.4	3
67	Novel Genetic Variants in Complement-Mediated Thrombotic Microangiopath. Blood, 2015, 126, 1050-1050.	1.4	3
68	Clinical Laboratory Immunology. American Journal of Clinical Pathology, 2014, 142, 437-444.	0.7	9
69	Newborn Screening for Severe Combined Immunodeficiency in 11 Screening Programs in the United States. JAMA - Journal of the American Medical Association, 2014, 312, 729.	7.4	586
70	Immune monitoring using the predictive power of immune profiles. , 2013, 1, 7.		50
71	Comprehensive immune monitoring reveals profound immunological changes in pancreas after kidney (PAK) transplant recipients. Human Immunology, 2013, 74, 738-745.	2.4	4
72	Adult-onset manifestation of idiopathic T-cell lymphopenia due to a heterozygous RAG1 mutation. Journal of Allergy and Clinical Immunology, 2013, 131, 1421-1423.	2.9	37

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73	Novel Infrastructure for Sepsis Biomarker Research in Critically Ill Neonates and Children. Clinical and Translational Science, 2013, 6, 21-25.	3.1	3
74	Atypical Omenn Syndrome due to Adenosine Deaminase Deficiency. Case Reports in Immunology, 2012, 2012, 1-4.	0.4	2
75	Relevance of antibody testing in patients with recurrent infections. Journal of Allergy and Clinical Immunology, 2012, 130, 558-559.e6.	2.9	9
76	SDF-1Î± Degrades whereas Glycoprotein 120 Upregulates Bcl-2 Interacting Mediator of Death Extralong Isoform: Implications for the Development of T Cell Memory. Journal of Immunology, 2012, 189, 1835-1842.	0.8	6
77	Association of an increased frequency of CD14⁺HLAâ€DR^{lo/neg} monocytes with decreased time to progression in chronic lymphocytic leukaemia (CLL). British Journal of Haematology, 2012, 156, 674-676.	2.5	58
78	Molecular Interrogation of Biclonal Multiple Myeloma for Clonal Relatedness.. Blood, 2012, 120, 2928-2928.	1.4	0
79	Utility of Doppler Myocardial Imaging, Cardiac Biomarkers, and Clonal Immunoglobulin Genes to Assess Left Ventricular Performance and Stratify Risk Following Peripheral Blood Stem Cell Transplantation in Patients with Systemic Light Chain Amyloidosis (AL). Journal of the American Society of Echocardiography, 2011, 24, 444-454.e2.	2.8	21
80	Immune evaluation and vaccine responses in Down syndrome: Evidence of immunodeficiency?. Vaccine, 2011, 29, 5040-5046.	3.8	39
81	Epidemiology of Cytomegalovirus Infection After Pancreas Transplantation. Transplantation, 2011, 92, 1044-1050.	1.0	22
82	A Reappraisal of Immunoglobulin Variable Gene Primers and Its Impact on Assessing Clonal Relationships Between PB B Cells and BM Plasma Cells in AL Amyloidosis. Journal of Clinical Immunology, 2011, 31, 1029-1037.	3.8	3
83	Accuracy of waste blood measurement in critically ill patients. Intensive Care Medicine, 2011, 37, 721-722.	8.2	3
84	Relevance of laboratory testing for the diagnosis of primary immunodeficiencies: a review of case-based examples of selected immunodeficiencies. Clinical and Molecular Allergy, 2011, 9, 6.	1.8	20
85	Epidemiology of Infections Requiring Hospitalization During Long-Term Follow-Up of Pancreas Transplantation. Transplantation, 2010, 89, 1126-1133.	1.0	38
86	Rituximab Therapy in Idiopathic Membranous Nephropathy. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 2188-2198.	4.5	247
87	Ig gene diversification and selection in follicular lymphoma, diffuse large B cell lymphoma and primary central nervous system lymphoma revealed by lineage tree and mutation analyses. International Immunology, 2010, 22, 875-887.	4.0	38
88	Phenotypic and clinical heterogeneity associated with monoallelic TNFRSF13Bâˆ’A181E mutations in common variable immunodeficiency. Human Immunology, 2010, 71, 505-511.	2.4	14
89	Proteomic Analysis of Immunoglobulin Light Chains (LC) In AL Amyloidosis Shows That the Sequence of Clonal LC Secreted by the Neoplastic Plasma Cells Is Identical to the LC Deposited In the Amyloid Plaques. Blood, 2010, 116, 1909-1909.	1.4	0
90	Immune Phenotyping and Naive T Cells as a Predictor of Response to Therapy In Chronic Lymphocytic Leukemia (CLL). Blood, 2010, 116, 1362-1362.	1.4	0

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91	Elevated serum immunoglobulin E (IgE): When to suspect hyper-IgE syndromeâ€"A 10-year pediatric tertiary care center experience. Allergy and Asthma Proceedings, 2009, 30, 23-27.	2.2	19
92	Common variable immunodeficiency: a new look at an old disease. Lancet, The, 2008, 372, 489-502.	13.7	289
93	Primary immunodeficiency: a call for multidisciplinary care â€“ Authors' reply. Lancet, The, 2008, 372, 1878.	13.7	0
94	Beneficial Effect of TRAIL on HIV Burden, without Detectable Immune Consequences. PLoS ONE, 2008, 3, e3096.	2.5	11
95	Serum-free light chainâ€"a new biomarker for patients with B-cell non-Hodgkin lymphoma and chronic lymphocytic leukemia. Translational Research, 2007, 149, 231-235.	5.0	53
96	Novel Analysis of Clonal Diversification in Blood B Cell and Bone Marrow Plasma Cell Clones in Immunoglobulin Light Chain Amyloidosis. Journal of Clinical Immunology, 2007, 27, 69-87.	3.8	18
97	Elimination of the Need for Urine Studies in the Screening Algorithm for Monoclonal Gammopathies by Using Serum Immunofixation and Free Light Chain Assays. Mayo Clinic Proceedings, 2006, 81, 1575-1578.	3.0	179
98	Absolute values of immunoglobulin free light chains are prognostic in patients with primary systemic amyloidosis undergoing peripheral blood stem cell transplantation. Blood, 2006, 107, 3378-3383.	1.4	230
99	Lineage tree analysis of immunoglobulin variable-region gene mutations in autoimmune diseases: Chronic activation, normal selection. Cellular Immunology, 2006, 244, 130-136.	3.0	20
100	Quantitative analysis of clonal bone marrow CD19+ B cells: Use of B cell lineage trees to delineate their role in the pathogenesis of light chain amyloidosis. Clinical Immunology, 2006, 120, 106-120.	3.2	13
101	Immunoglobulin variable-region gene mutational lineage tree analysis: Application to autoimmune diseases. Autoimmunity Reviews, 2006, 5, 242-251.	5.8	15
102	Bence Jones Cryoglobulinuria: Characterization of a Urinary Îº Light Chain Cryoglobulin. Clinical Chemistry, 2006, 52, 1435-1436.	3.2	5
103	Functional gene expression analysis of clonal plasma cells identifies a unique molecular profile for light chain amyloidosis. Blood, 2005, 105, 794-803.	1.4	52
104	Diagnostic Performance of Quantitative Îº and Î» Free Light Chain Assays in Clinical Practice. Clinical Chemistry, 2005, 51, 878-881.	3.2	244
105	Absolute Values of Serum Immunoglobulin Free Light Chains Predict for Survival in Patients with Primary Systemic Amyloidosis Undergoing Peripheral Blood Stem Cell Transplant.. Blood, 2005, 106, 422-422.	1.4	0
106	Analysis of Somatic Hypermutation and Antigenic Selection in the Clonal B Cell in Immunoglobulin Light Chain Amyloidosis (AL). Journal of Clinical Immunology, 2004, 24, 340-353.	3.8	24
107	Characterization of amyloidogenic immunoglobulin light chains directly from serum by on-line immunoafï¬nity isolation. Biomedical Chromatography, 2004, 18, 191-201.	1.7	19
108	Quantitative Analysis of Serum Free Light Chains. American Journal of Clinical Pathology, 2003, 119, 274-278.	0.7	176

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109	Immunoglobulin light chain variable (V) region genes influence clinical presentation and outcome in light chainâ€“associated amyloidosis (AL). Blood, 2003, 101, 3801-3807.	1.4	207
110	Modulation of insulitis and type 1 diabetes by transgenic HLA-DR3 and DQ8 in NOD mice lacking endogenous MHC class II. Human Immunology, 2002, 63, 987-999.	2.4	23
111	Correlation of Serum Immunoglobulin Free Light Chain Quantification with Urinary Bence Jones Protein in Light Chain Myeloma. Clinical Chemistry, 2002, 48, 655-657.	3.2	115
112	HLA-DQ8 transgenic and NOD mice recognize different epitopes within the cytoplasmic region of the tyrosine phosphatase-like molecule, IA-2. Human Immunology, 2001, 62, 1099-1105.	2.4	21
113	Type 1 Diabetes-Predisposing MHC Alleles Influence the Selection of Glutamic Acid Decarboxylase (GAD) 65-Specific T Cells in a Transgenic Model. Journal of Immunology, 2001, 166, 1370-1379.	0.8	30
114	Identification of HLA-class-II-restricted epitopes of autoantigens in transgenic mice. Current Opinion in Immunology, 2000, 12, 122-129.	5.5	28
115	NOD background genes influence T cell responses to GAD 65 in HLA-DQ8 transgenic mice. Human Immunology, 1999, 60, 583-590.	2.4	24