

# 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	Newborn Screening for Severe Combined Immunodeficiency in 11 Screening Programs in the United States. <i>JAMA - Journal of the American Medical Association</i> , 2014, 312, 729.	7.4	586
2	Common variable immunodeficiency: a new look at an old disease. <i>Lancet, The</i> , 2008, 372, 489-502.	13.7	289
3	Coronavirus disease 2019 in patients with inborn errors of immunity: An international study. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 520-531.	2.9	278
4	Rituximab Therapy in Idiopathic Membranous Nephropathy. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2010, 5, 2188-2198.	4.5	247
5	Diagnostic Performance of Quantitative $\kappa$ and $\lambda$ Free Light Chain Assays in Clinical Practice. <i>Clinical Chemistry</i> , 2005, 51, 878-881.	3.2	244
6	Absolute values of immunoglobulin free light chains are prognostic in patients with primary systemic amyloidosis undergoing peripheral blood stem cell transplantation. <i>Blood</i> , 2006, 107, 3378-3383.	1.4	230
7	Immunoglobulin light chain variable (V) region genes influence clinical presentation and outcome in light chain-associated amyloidosis (AL). <i>Blood</i> , 2003, 101, 3801-3807.	1.4	207
8	Elimination of the Need for Urine Studies in the Screening Algorithm for Monoclonal Gammopathies by Using Serum Immunofixation and Free Light Chain Assays. <i>Mayo Clinic Proceedings</i> , 2006, 81, 1575-1578.	3.0	179
9	Quantitative Analysis of Serum Free Light Chains. <i>American Journal of Clinical Pathology</i> , 2003, 119, 274-278.	0.7	176
10	Broad-spectrum antibodies against self-antigens and cytokines in RAG deficiency. <i>Journal of Clinical Investigation</i> , 2015, 125, 4135-4148.	8.2	159
11	Correlation of Serum Immunoglobulin Free Light Chain Quantification with Urinary Bence Jones Protein in Light Chain Myeloma. <i>Clinical Chemistry</i> , 2002, 48, 655-657.	3.2	115
12	Consequences of B-cell-depleting therapy: hypogammaglobulinemia and impaired B-cell reconstitution. <i>Immunotherapy</i> , 2018, 10, 713-728.	2.0	93
13	Use of Genetic Testing for Primary Immunodeficiency Patients. <i>Journal of Clinical Immunology</i> , 2018, 38, 320-329.	3.8	88
14	Outcome of Whole Exome Sequencing for Diagnostic Odyssey Cases of an Individualized Medicine Clinic. <i>Mayo Clinic Proceedings</i> , 2016, 91, 297-307.	3.0	83
15	Short telomere syndromes cause a primary T cell immunodeficiency. <i>Journal of Clinical Investigation</i> , 2018, 128, 5222-5234.	8.2	82
16	Flow Cytometry, a Versatile Tool for Diagnosis and Monitoring of Primary Immunodeficiencies. <i>Vaccine Journal</i> , 2016, 23, 254-271.	3.1	76
17	Dermatologic Features of ADA2 Deficiency in Cutaneous Polyarteritis Nodosa. <i>JAMA Dermatology</i> , 2015, 151, 1230.	4.1	75
18	Outcomes and Treatment Strategies for Autoimmunity and Hyperinflammation in Patients with RAG Deficiency. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2019, 7, 1970-1985.e4.	3.8	64

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19	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). <i>Frontiers in Immunology</i> , 2019, 10, 777.	4.8	59
20	Association of an increased frequency of CD14 <sup>+</sup> HLA-DR <sup>lo/neg</sup> monocytes with decreased time to progression in chronic lymphocytic leukaemia (CLL). <i>British Journal of Haematology</i> , 2012, 156, 674-676.	2.5	58
21	Thrombotic Microangiopathy Care Pathway: A Consensus Statement for the Mayo Clinic Complement Alternative Pathway-Thrombotic Microangiopathy (CAP-TMA) Disease-Oriented Group. <i>Mayo Clinic Proceedings</i> , 2016, 91, 1189-1211.	3.0	55
22	PAX1 is essential for development and function of the human thymus. <i>Science Immunology</i> , 2020, 5, .	11.9	55
23	Serum-free light chain—a new biomarker for patients with B-cell non-Hodgkin lymphoma and chronic lymphocytic leukemia. <i>Translational Research</i> , 2007, 149, 231-235.	5.0	53
24	Functional gene expression analysis of clonal plasma cells identifies a unique molecular profile for light chain amyloidosis. <i>Blood</i> , 2005, 105, 794-803.	1.4	52
25	Gastrointestinal Manifestations in X-linked Agammaglobulinemia. <i>Journal of Clinical Immunology</i> , 2017, 37, 287-294.	3.8	51
26	Immune monitoring using the predictive power of immune profiles. , 2013, 1, 7.		50
27	Activating mutations in PIK3CD disrupt the differentiation and function of human and murine CD4+ T cells. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 236-253.	2.9	44
28	Spectrum of myeloid neoplasms and immune deficiency associated with germline <i>GATA2</i> mutations. <i>Cancer Medicine</i> , 2015, 4, 490-499.	2.8	43
29	Immune evaluation and vaccine responses in Down syndrome: Evidence of immunodeficiency?. <i>Vaccine</i> , 2011, 29, 5040-5046.	3.8	39
30	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. <i>American Journal of Human Genetics</i> , 2019, 105, 108-121.	6.2	39
31	Epidemiology of Infections Requiring Hospitalization During Long-Term Follow-Up of Pancreas Transplantation. <i>Transplantation</i> , 2010, 89, 1126-1133.	1.0	38
32	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. <i>International Immunology</i> , 2010, 22, 875-887.	4.0	38
33	Adult-onset manifestation of idiopathic T-cell lymphopenia due to a heterozygous RAG1 mutation. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 131, 1421-1423.	2.9	37
34	Type 1 Diabetes-Predisposing MHC Alleles Influence the Selection of Glutamic Acid Decarboxylase (GAD) 65-Specific T Cells in a Transgenic Model. <i>Journal of Immunology</i> , 2001, 166, 1370-1379.	0.8	30
35	Utility of DNA, RNA, Protein, and Functional Approaches to Solve Cryptic Immunodeficiencies. <i>Journal of Clinical Immunology</i> , 2018, 38, 307-319.	3.8	29
36	Clinical Correlation of Cytomegalovirus Infection With CMV-specific CD8+ T-cell Immune Competence Score and Lymphocyte Subsets in Solid Organ Transplant Recipients. <i>Transplantation</i> , 2019, 103, 832-838.	1.0	29

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37	Identification of HLA-class-II-restricted epitopes of autoantigens in transgenic mice. <i>Current Opinion in Immunology</i> , 2000, 12, 122-129.	5.5	28
38	Combinatorial Immunoprofiling in Latent Tuberculosis Infection. Toward Better Risk Stratification. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2015, 192, 605-617.	5.6	28
39	A Novel Pathogenic Variant in CARMIL2 (RLTPR) Causing CARMIL2 Deficiency and EBV-Associated Smooth Muscle Tumors. <i>Frontiers in Immunology</i> , 2020, 11, 884.	4.8	26
40	Diagnostic accuracy and clinical relevance of an inflammatory biomarker panel for sepsis in adult critically ill patients. <i>Diagnostic Microbiology and Infectious Disease</i> , 2016, 84, 175-180.	1.8	25
41	Severe SARS-CoV-2 disease in the context of a NF- $\kappa$ B2 loss-of-function pathogenic variant. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 532-544.e1.	2.9	25
42	NOD background genes influence T cell responses to GAD 65 in HLA-DQ8 transgenic mice. <i>Human Immunology</i> , 1999, 60, 583-590.	2.4	24
43	Analysis of Somatic Hypermutation and Antigenic Selection in the Clonal B Cell in Immunoglobulin Light Chain Amyloidosis (AL). <i>Journal of Clinical Immunology</i> , 2004, 24, 340-353.	3.8	24
44	Modulation of insulinitis and type 1 diabetes by transgenic HLA-DR3 and DQ8 in NOD mice lacking endogenous MHC class II. <i>Human Immunology</i> , 2002, 63, 987-999.	2.4	23
45	Monocyte HLA-DR expression and neutrophil CD64 expression as biomarkers of infection in critically ill neonates and infants. <i>Pediatric Research</i> , 2015, 78, 683-690.	2.3	23
46	Epidemiology of Cytomegalovirus Infection After Pancreas Transplantation. <i>Transplantation</i> , 2011, 92, 1044-1050.	1.0	22
47	HLA-DQ8 transgenic and NOD mice recognize different epitopes within the cytoplasmic region of the tyrosine phosphatase-like molecule, IA-2. <i>Human Immunology</i> , 2001, 62, 1099-1105.	2.4	21
48	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). <i>Journal of the American Society of Echocardiography</i> , 2011, 24, 444-454.e2.	2.8	21
49	Lineage tree analysis of immunoglobulin variable-region gene mutations in autoimmune diseases: Chronic activation, normal selection. <i>Cellular Immunology</i> , 2006, 244, 130-136.	3.0	20
50	Relevance of laboratory testing for the diagnosis of primary immunodeficiencies: a review of case-based examples of selected immunodeficiencies. <i>Clinical and Molecular Allergy</i> , 2011, 9, 6.	1.8	20
51	Clinical Correlates and Treatment Outcomes for Patients With Short Telomere Syndromes. <i>Mayo Clinic Proceedings</i> , 2018, 93, 834-839.	3.0	20
52	Characterization of amyloidogenic immunoglobulin light chains directly from serum by on-line immunoaffinity isolation. <i>Biomedical Chromatography</i> , 2004, 18, 191-201.	1.7	19
53	Elevated serum immunoglobulin E (IgE): When to suspect hyper-IgE syndrome? A 10-year pediatric tertiary care center experience. <i>Allergy and Asthma Proceedings</i> , 2009, 30, 23-27.	2.2	19
54	Unrelated Hematopoietic Cell Transplantation in a Patient with Combined Immunodeficiency with Granulomatous Disease and Autoimmunity Secondary to RAG Deficiency. <i>Journal of Clinical Immunology</i> , 2016, 36, 725-732.	3.8	19

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55	Rubella Virus Infected Macrophages and Neutrophils Define Patterns of Granulomatous Inflammation in Inborn and Acquired Errors of Immunity. <i>Frontiers in Immunology</i> , 2021, 12, 796065.	4.8	19
56	Novel Analysis of Clonal Diversification in Blood B Cell and Bone Marrow Plasma Cell Clones in Immunoglobulin Light Chain Amyloidosis. <i>Journal of Clinical Immunology</i> , 2007, 27, 69-87.	3.8	18
57	Diagnostic Utility of Complement Serology for Atypical Hemolytic Uremic Syndrome. <i>Mayo Clinic Proceedings</i> , 2018, 93, 1351-1362.	3.0	17
58	WILD syndrome is GATA2 deficiency: A novel deletion in the GATA2 gene. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2017, 5, 1149-1152.e1.	3.8	16
59	Immunoglobulin variable-region gene mutational lineage tree analysis: Application to autoimmune diseases. <i>Autoimmunity Reviews</i> , 2006, 5, 242-251.	5.8	15
60	Diagnostic Tools for Inborn Errors of Human Immunity (Primary Immunodeficiencies and Immune) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	5.3	15
61	The total IgM, IgA and IgG antibody responses to pneumococcal polysaccharide vaccination (Pneumovax®23) in a healthy adult population and patients diagnosed with primary immunodeficiencies. <i>Vaccine</i> , 2019, 37, 1350-1355.	3.8	15
62	Phenotypic and clinical heterogeneity associated with monoallelic TNFRSF13B A181E mutations in common variable immunodeficiency. <i>Human Immunology</i> , 2010, 71, 505-511.	2.4	14
63	A Droplet Digital PCR Method for Severe Combined Immunodeficiency Newborn Screening. <i>Journal of Molecular Diagnostics</i> , 2017, 19, 755-765.	2.8	14
64	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. <i>Clinical Immunology</i> , 2006, 120, 106-120.	3.2	13
65	Application of a radiosensitivity flow assay in a patient with DNA ligase 4 deficiency. <i>Blood Advances</i> , 2018, 2, 1828-1832.	5.2	13
66	Follow-Up for an Abnormal Newborn Screen for Severe Combined Immunodeficiencies (NBS SCID): A Clinical Immunology Society (CIS) Survey of Current Practices. <i>International Journal of Neonatal Screening</i> , 2020, 6, 52.	3.2	12
67	Next-generation sequencing for inborn errors of immunity. <i>Human Immunology</i> , 2021, 82, 871-882.	2.4	12
68	Beneficial Effect of TRAIL on HIV Burden, without Detectable Immune Consequences. <i>PLoS ONE</i> , 2008, 3, e3096.	2.5	11
69	Successful Nonmyeloablative Allogeneic Stem Cell Transplant in a Child With Emberger Syndrome and GATA2 Mutation. <i>Journal of Pediatric Hematology/Oncology</i> , 2018, 40, e383-e388.	0.6	10
70	How to evaluate for immunodeficiency in patients with autoimmune cytopenias: laboratory evaluation for the diagnosis of inborn errors of immunity associated with immune dysregulation. <i>Hematology American Society of Hematology Education Program</i> , 2020, 2020, 661-672.	2.5	10
71	POLD1 Deficiency Reveals a Role for POLD1 in DNA Repair and T and B Cell Development. <i>Journal of Clinical Immunology</i> , 2021, 41, 270-273.	3.8	10
72	The New "Wholly Trinity" in the Diagnosis and Management of Inborn Errors of Immunity. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 613-625.	3.8	10

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73	Defining the mild variant of leukocyte adhesion deficiency type 2 (SLC35C1-congenital disorder of glycosylation) and response to enzyme replacement therapy: Insights from two new families and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2005-2018.	1.2	10
74	Relevance of antibody testing in patients with recurrent infections. <i>Journal of Allergy and Clinical Immunology</i> , 2012, 130, 558-559.e6.	2.9	9
75	Clinical Laboratory Immunology. <i>American Journal of Clinical Pathology</i> , 2014, 142, 437-444.	0.7	9
76	Relationship between asthma status and antibody response pattern to 23-valent pneumococcal vaccination. <i>Journal of Asthma</i> , 2020, 57, 381-390.	1.7	8
77	NADPH Oxidase-Specific Flow Cytometry Allows for Rapid Genetic Triage and Classification of Novel Variants in Chronic Granulomatous Disease. <i>Journal of Clinical Immunology</i> , 2020, 40, 191-202.	3.8	8
78	Functional Confirmation of DNA Repair Defect in Ataxia Telangiectasia (AT) Infants Identified by Newborn Screening for Severe Combined Immunodeficiency (NBS SCID). <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 723-732.e3.	3.8	8
79	Defective TLR9-driven STAT3 activation in B cells of patients with COVID-19. <i>Clinical Immunology</i> , 2018, 197, 40-44.	3.2	7
80	Pneumococcal serotype-specific cut-offs based on antibody responses to pneumococcal polysaccharide vaccination in healthy adults. <i>Vaccine</i> , 2021, 39, 2850-2856.	3.8	7
81	SDF-1 $\alpha$ Degrades whereas Glycoprotein 120 Upregulates Bcl-2 Interacting Mediator of Death Extralong Isoform: Implications for the Development of T Cell Memory. <i>Journal of Immunology</i> , 2012, 189, 1835-1842.	0.8	6
82	RNA sequencing reveals the consequences of a novel insertion in dedicator of cytokinesis-8. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 289-292.e6.	2.9	6
83	A case of atypical, complete DiGeorge syndrome without 22q11.2 deletion. <i>Annals of Allergy, Asthma and Immunology</i> , 2017, 118, 640-642.e2.	1.0	6
84	X-Linked Lymphoproliferative Syndrome Presenting as Adult-Onset Multi-Infarct Dementia. <i>Journal of Neuropathology and Experimental Neurology</i> , 2019, 78, 460-466.	1.7	6
85	Bence Jones Cryoglobulinuria: Characterization of a Urinary $\kappa$ Light Chain Cryoglobulin. <i>Clinical Chemistry</i> , 2006, 52, 1435-1436.	3.2	5
86	Comprehensive immune monitoring reveals profound immunological changes in pancreas after kidney (PAK) transplant recipients. <i>Human Immunology</i> , 2013, 74, 738-745.	2.4	4
87	Clinical spectrum and clonal evolution in germline syndromes with predisposition to myeloid neoplasms. <i>British Journal of Haematology</i> , 2018, 182, 141-145.	2.5	4
88	Phenotypic heterogeneity associated with germline <i>GATA2</i> haploinsufficiency: a comprehensive kindred study. <i>Leukemia and Lymphoma</i> , 2019, 60, 3282-3286.	1.3	4
89	Editorial: Application of Cytometry in Primary Immunodeficiencies. <i>Frontiers in Immunology</i> , 2020, 11, 463.	4.8	4
90	Case report and review of the literature: immune dysregulation in a large familial cohort due to a novel pathogenic <i>RELA</i> variant. <i>Rheumatology</i> , 2022, 62, 347-359.	1.9	4

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91	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. <i>Journal of Clinical Immunology</i> , 2011, 31, 1029-1037.	3.8	3
92	Accuracy of waste blood measurement in critically ill patients. <i>Intensive Care Medicine</i> , 2011, 37, 721-722.	8.2	3
93	Novel Infrastructure for Sepsis Biomarker Research in Critically Ill Neonates and Children. <i>Clinical and Translational Science</i> , 2013, 6, 21-25.	3.1	3
94	Provider Perceptions of Quality of Life, Neurocognition, Physical Well-being, and Psychosocial Health in Patients with Primary Immunodeficiency/Immune Dysregulation Conditions. <i>Journal of Clinical Immunology</i> , 2019, 39, 805-813.	3.8	3
95	Delayed-Onset ADA1 (ADA) Deficiency Not Detected by TREC Screen. <i>Pediatrics</i> , 2021, 147, e2020005579.	2.1	3
96	Do monogenic inborn errors of immunity cause susceptibility to severe COVID-19?. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	3
97	Sex differences in antibody responses to the 23-valent pneumococcal polysaccharide vaccination. <i>Annals of Allergy, Asthma and Immunology</i> , 2021, 127, 509-510.	1.0	3
98	Cryptococcal pneumonia in an adolescent with a gain-of-function variant in signal transduction and activator of transcription 1 ( <i>STAT1</i> ). <i>BMJ Case Reports</i> , 2020, 13, e234120.	0.5	3
99	Analysis of Serum Ferritin Levels As a Diagnostic Criteria for Hemophagocytic Lymphohistiocytosis (HLH) in Hospitalized Adult Patients. <i>Blood</i> , 2015, 126, 1014-1014.	1.4	3
100	Novel Genetic Variants in Complement-Mediated Thrombotic Microangiopath. <i>Blood</i> , 2015, 126, 1050-1050.	1.4	3
101	Atypical Omenn Syndrome due to Adenosine Deaminase Deficiency. <i>Case Reports in Immunology</i> , 2012, 2012, 1-4.	0.4	2
102	Optimal approach to assessing T-cell function in haematopoietic cell transplant recipients. <i>BMJ Case Reports</i> , 2018, 2018, bcr-2017-222417.	0.5	2
103	“The State of the Union”: Current and Future Perspectives on Patient-Centric Care for Primary Immunodeficiencies and Immune Dysregulatory Diseases. <i>Frontiers in Immunology</i> , 2019, 10, 1783.	4.8	2
104	Infectious Complications Predict Premature CD8+ T-cell Senescence in CD40 Ligand-Deficient Patients. <i>Journal of Clinical Immunology</i> , 2021, 41, 795-806.	3.8	2
105	Immune Reconstitution after Hematopoietic Stem Cell Transplantation in Immunodeficiency“Centromeric Instability“Facial Anomalies Syndrome Type 1. <i>Journal of Clinical Immunology</i> , 2021, 41, 1089-1094.	3.8	2
106	Identification of 22 novel BTK gene variants in B cell deficiency with hypogammaglobulinemia. <i>Clinical Immunology</i> , 2021, 229, 108788.	3.2	2
107	Newborn Screening for Severe Combined Immunodeficiency: Changing the Landscape of Pediatric Primary Immunodeficiencies. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2015, 3, 1008-1009.	3.8	1
108	Expanding mechanistic insights into the pathogenesis of idiopathic CD4+ T cell lymphocytopenia. <i>Journal of Clinical Investigation</i> , 2020, 130, 5105-5108.	8.2	1

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109	Supporting Careers of Women in Clinical Immunology: From Conceptualization to Implementation. <i>Frontiers in Pediatrics</i> , 2022, 10, 864734.	1.9	1
110	Primary immunodeficiency: a call for multidisciplinary care – Authors' reply. <i>Lancet</i> , The, 2008, 372, 1878.	13.7	0
111	Absolute Values of Serum Immunoglobulin Free Light Chains Predict for Survival in Patients with Primary Systemic Amyloidosis Undergoing Peripheral Blood Stem Cell Transplant.. <i>Blood</i> , 2005, 106, 422-422.	1.4	0
112	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. <i>Blood</i> , 2010, 116, 1909-1909.	1.4	0
113	Immune Phenotyping and Naive T Cells as a Predictor of Response to Therapy In Chronic Lymphocytic Leukemia (CLL). <i>Blood</i> , 2010, 116, 1362-1362.	1.4	0
114	Molecular Interrogation of Biclinal Multiple Myeloma for Clonal Relatedness.. <i>Blood</i> , 2012, 120, 2928-2928.	1.4	0
115	Interpretation of Dihydrorhodamine-1,2,3 Flow Cytometry in Chronic Granulomatous Disease: an Atypical Exemplar. <i>Journal of Clinical Immunology</i> , 2022, , 1.	3.8	0