Janet Chou

List of Publications by Year
in descending order

93 papers	6,056 citations	87888 38 h-index	76900 74 g-index
96 all docs	96 docs citations	96 times ranked	8444 citing authors

#	Article	IF	Citations
1	Advances in clinical outcomes: What we have learned during the COVID-19 pandemic. Journal of Allergy and Clinical Immunology, 2022, 149, 569-578.	2.9	3
2	Immunology of SARS-CoV-2 infection in children. Nature Immunology, 2022, 23, 177-185.	14.5	102
3	An adjuvant strategy enabled by modulation of the physical properties of microbial ligands expands antigen immunogenicity. Cell, 2022, 185, 614-629.e21.	28.9	40
4	Multisystem Inflammatory-like Syndrome in a Child Following COVID-19 mRNA Vaccination. Vaccines, 2022, 10, 43.	4.4	21
5	Genetic diagnosis of immune dysregulation can lead to targeted therapy for interstitial lung disease: A case series and single center approach. Pediatric Pulmonology, 2022, 57, 1577-1587.	2.0	4
6	ITK deficiency presenting as autoimmune lymphoproliferative syndrome. Journal of Allergy and Clinical Immunology, 2021, 147, 743-745.e1.	2.9	8
7	Hematopoietic Stem Cell Transplantation Is a Curative Therapy for Transferrin Receptor 1 (TFRC) Deficiency. Journal of Allergy and Clinical Immunology: in Practice, 2021, 9, 753-759.e2.	3.8	4
8	Efficacy and economics of targeted panel versus whole-exome sequencing in 878 patients with suspected primary immunodeficiency. Journal of Allergy and Clinical Immunology, 2021, 147, 723-726.	2.9	31
9	Combined immunodeficiency due to a mutation in the Î³1 subunit of the coat protein I complex. Journal of Clinical Investigation, 2021, 131, .	8.2	15
10	Thymopoiesis, Alterations in Dendritic Cells and Tregs, and Reduced T Cell Activation in Successful Extracorporeal Photopheresis Treatment of GVHD. Journal of Clinical Immunology, 2021, 41, 1016-1030.	3.8	9
11	Multisystem inflammation and susceptibility to viral infections in human ZNFX1 deficiency. Journal of Allergy and Clinical Immunology, 2021, 148, 381-393.	2.9	40
12	The interferon landscape along the respiratory tract impacts the severity of COVID-19. Cell, 2021, 184, 4953-4968.e16.	28.9	165
13	Mechanisms underlying genetic susceptibility to multisystem inflammatory syndrome in children (MIS-C). Journal of Allergy and Clinical Immunology, 2021, 148, 732-738.e1.	2.9	84
14	Combined immunodeficiency with autoimmunity caused by a homozygous missense mutation in inhibitor of nuclear factor ?B kinase alpha (IKKÎ±). Science Immunology, 2021, 6, eabf6723.	11.9	6
15	Expanding the Nude SCID/CID Phenotype Associated with FOXN1 Homozygous, Compound Heterozygous, or Heterozygous Mutations. Journal of Clinical Immunology, 2021, 41, 756-768.	3.8	13
16	Diagnostic interpretation of genetic studies in patients with primary immunodeficiency diseases: AÂworking group report of the Primary Immunodeficiency Diseases Committee of the American Academy of Allergy, Asthma & Immunology. Journal of Allergy and Clinical Immunology, 2020, 145, 46-69.	2.9	54
17	Hypomorphic variants in AK2 reveal the contribution of mitochondrial function to B-cell activation. Journal of Allergy and Clinical Immunology, 2020, 146, 192-202.	2.9	13
18	Dysregulated actin dynamics in activated PI3KÎ´ syndrome. Clinical Immunology, 2020, 210, 108311.	3.2	7

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19	Acetaminophen Inhibits the Neutrophil Oxidative Burst: Implications for Diagnostic Testing. Journal of Allergy and Clinical Immunology: in Practice, 2020, 8, 3543-3548.	3.8	0
20	Immune dysregulation and multisystem inflammatory syndrome in children (MIS-C) in individuals with haploinsufficiency of SOCS1. Journal of Allergy and Clinical Immunology, 2020, 146, 1194-1200.e1.	2.9	92
21	A Case of STK4 Deficiency with Complications Evoking Mycobacterial Infection. Journal of Clinical Immunology, 2020, 40, 665-669.	3.8	8
22	Genotype and functional correlates of disease phenotype in deficiency of adenosine deaminase 2 (DADA2). Journal of Allergy and Clinical Immunology, 2020, 145, 1664-1672.e10.	2.9	95
23	Severe combined immunodeficiency caused by inositol-trisphosphate 3-kinase B (ITPKB) deficiency. Journal of Allergy and Clinical Immunology, 2020, 145, 1696-1699.e6.	2.9	6
24	Autoimmunity and immunodeficiency. Current Opinion in Rheumatology, 2020, 32, 168-174.	4.3	10
25	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
26	LRRC8 family proteins within lysosomes regulate cellular osmoregulation and enhance cell survival to multiple physiological stresses. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 29155-29165.	7.1	36
27	Distinct clinical and immunological features of SARSâ€“CoV-2â€“induced multisystem inflammatory syndrome in children. Journal of Clinical Investigation, 2020, 130, 5942-5950.	8.2	287
28	A novel truncating mutation in MYD88 in a patient with BCG adenitis, neutropenia and delayed umbilical cord separation. Clinical Immunology, 2019, 207, 40-42.	3.2	9
29	Combined immunodeficiency in a patient with c-Rel deficiency. Journal of Allergy and Clinical Immunology, 2019, 144, 606-608.e4.	2.9	32
30	Rethinking newborn screening for severe combined immunodeficiency: Lessons from an international partnership for patients with primary immunodeficiencies in Pakistan. Clinical Immunology, 2019, 202, 29-32.	3.2	2
31	T-cell mitochondrial dysfunction and lymphopenia in DOCK2-deficient patients. Journal of Allergy and Clinical Immunology, 2019, 144, 306-309.e2.	2.9	13
32	F-BAR domain only protein 1 (FCHO1) deficiency is a novel cause of combined immune deficiency in human subjects. Journal of Allergy and Clinical Immunology, 2019, 143, 2317-2321.e12.	2.9	21
33	Immunodeficiency and EBV-induced lymphoproliferation caused by 4-1BB deficiency. Journal of Allergy and Clinical Immunology, 2019, 144, 574-583.e5.	2.9	63
34	Human primary immunodeficiency caused by expression of a kinase-dead p110Î´ mutant. Journal of Allergy and Clinical Immunology, 2019, 143, 797-799.e2.	2.9	33
35	Immunologic reconstitution following hematopoietic stem cell transplantation despite lymph node paucity in NF-Î²Bâ€“inducing kinase deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 1240-1243.e4.	2.9	6
36	Atypical phenotype of an old disease or typical phenotype of a new disease: deficiency of adenosine deaminase 2. Turkish Journal of Pediatrics, 2019, 61, 413.	0.6	5

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37	A young girl with severe cerebral fungal infection due to card 9 deficiency. Clinical Immunology, 2018, 191, 21-26.	3.2	27
38	Exaggerated follicular helper T-cell responses in patients with LRBA deficiency caused by failure of CTLA4-mediated regulation. Journal of Allergy and Clinical Immunology, 2018, 141, 1050-1059.e10.	2.9	93
39	DNA recombination defects in Kuwait: Clinical, immunologic and genetic profile. Clinical Immunology, 2018, 187, 68-75.	3.2	11
40	Clinical, immunologic, and genetic spectrum of 696 patients with combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2018, 141, 1450-1458.	2.9	90
41	Novel biallelic TRNT1 mutations resulting in sideroblastic anemia, combined B and T cell defects, hypogammaglobulinemia, recurrent infections, hypertrophic cardiomyopathy and developmental delay. Clinical Immunology, 2018, 188, 20-22.	3.2	24
42	Phenotype, penetrance, and treatment of 133 cytotoxic T-lymphocyte antigen 4â€“insufficient subjects. Journal of Allergy and Clinical Immunology, 2018, 142, 1932-1946.	2.9	344
43	A recessive form of hyper-IgE syndrome by disruption of ZNF341-dependent STAT3 transcription and activity. Science Immunology, 2018, 3, .	11.9	132
44	Comprehensive Genetic Results for Primary Immunodeficiency Disorders in a Highly Consanguineous Population. Frontiers in Immunology, 2018, 9, 3146.	4.8	37
45	Deficient LRRC8A-dependent volume-regulated anion channel activity is associated with male infertility in mice. JCI Insight, 2018, 3, .	5.0	29
46	Leucine-rich repeat containing 8A (LRRC8A) â€“dependent volume-regulated anion channel activity is dispensable for T-cell development and function. Journal of Allergy and Clinical Immunology, 2017, 140, 1651-1659.e1.	2.9	36
47	Human RELA haploinsufficiency results in autosomal-dominant chronic mucocutaneous ulceration. Journal of Experimental Medicine, 2017, 214, 1937-1947.	8.5	84
48	DOCK8 Deficiency Presenting as an IPEX-Like Disorder. Journal of Clinical Immunology, 2017, 37, 811-819.	3.8	39
49	Detection of Sp110 by Flow Cytometry and Application to Screening Patients for Veno-occlusive Disease with Immunodeficiency. Journal of Clinical Immunology, 2017, 37, 707-714.	3.8	11
50	Combined immunodeficiency with EBV positive B cell lymphoma and epidermodysplasia verruciformis due to a novel homozygous mutation in RASGRP1. Clinical Immunology, 2017, 183, 142-144.	3.2	43
51	A novel mutation in the JH4 domain of JAK3 causing severe combined immunodeficiency complicated by vertebral osteomyelitis. Clinical Immunology, 2017, 183, 198-200.	3.2	6
52	Cernunnos deficiency associated with BCG adenitis and autoimmunity: First case from the national Iranian registry and review of the literature. Clinical Immunology, 2017, 183, 201-206.	3.2	11
53	Epidermodysplasia verruciformis as a manifestation of ARTEMIS deficiency in a young adult. Journal of Allergy and Clinical Immunology, 2017, 139, 372-375.e4.	2.9	18
54	Long-term outcomes of 176 patients with X-linked hyper-IgM syndrome treated with or without hematopoietic cell transplantation. Journal of Allergy and Clinical Immunology, 2017, 139, 1282-1292.	2.9	107

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55	The LRRC8A Mediated â€œSwell Activatedâ€•Chloride Conductance Is Dispensable for Vacuolar Homeostasis in Neutrophils. Frontiers in Pharmacology, 2017, 8, 262.	3.5	9
56	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
57	Uses of Next-Generation Sequencing Technologies for the Diagnosis of Primary Immunodeficiencies. Frontiers in Immunology, 2017, 8, 847.	4.8	95
58	14â€‰Years after Discovery: Clinical Follow-up on 15 Patients with Inducible Co-Stimulator Deficiency. Frontiers in Immunology, 2017, 8, 964.	4.8	57
59	A digenic human immunodeficiency characterized by IFNAR1 and IFNGR2 mutations. Journal of Clinical Investigation, 2017, 127, 4415-4420.	8.2	53
60	Chronic mucocutaneous candidiasis associated with an SH2 domain gain-of-function mutation that enhances STAT1 phosphorylation. Journal of Allergy and Clinical Immunology, 2016, 138, 297-299.	2.9	24
61	Hematopoietic stem cell transplantation outcomes for 11 patients with dedicator of cytokinesis 8 deficiency. Journal of Allergy and Clinical Immunology, 2016, 138, 852-859.e3.	2.9	48
62	Combined immunodeficiency due to a homozygous mutation in ORAI1 that deletes the C-terminus that interacts with STIM 1. Clinical Immunology, 2016, 166-167, 100-102.	3.2	11
63	Mutations in pyrin masquerading as a primary immunodeficiency. Clinical Immunology, 2016, 171, 65-66.	3.2	2
64	A missense mutation in TFRC, encoding transferrin receptor 1, causes combined immunodeficiency. Nature Genetics, 2016, 48, 74-78.	21.4	219
65	Spectrum of Phenotypes Associated with Mutations in LRBA. Journal of Clinical Immunology, 2016, 36, 33-45.	3.8	180
66	Recurrent viral infections associated with a homozygous CORO1A mutation that disrupts oligomerization and cytoskeletal association. Journal of Allergy and Clinical Immunology, 2016, 137, 879-888.e2.	2.9	41
67	Autoimmune lymphoproliferative syndrome caused by a homozygous FasL mutation that disrupts FasL assembly. Journal of Allergy and Clinical Immunology, 2016, 137, 324-327.e2.	2.9	13
68	Deficiency of base excision repair enzyme NEIL3 drives increased predisposition to autoimmunity. Journal of Clinical Investigation, 2016, 126, 4219-4236.	8.2	56
69	Inherited DOCK2 Deficiency in Patients with Early-Onset Invasive Infections. New England Journal of Medicine, 2015, 372, 2409-2422.	27.0	169
70	A novel mutation in NCF2 associated with autoimmune disease and a solitary late-onset infection. Clinical Immunology, 2015, 161, 128-130.	3.2	12
71	A novel mutation in ORAI1 presenting with combined immunodeficiency and residual T-cell function. Journal of Allergy and Clinical Immunology, 2015, 136, 479-482.e1.	2.9	28
72	Broad spectrum of autoantibodies in patients with Wiskott-Aldrich syndrome and X-linked thrombocytopenia. Journal of Allergy and Clinical Immunology, 2015, 136, 1401-1404.e3.	2.9	25

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73	A novel mutation in ICOS presenting as hypogammaglobulinemia with susceptibility to opportunistic pathogens. Journal of Allergy and Clinical Immunology, 2015, 136, 794-797.e1.	2.9	26
74	Regulatory T-cell deficiency and immune dysregulation, polyendocrinopathy, enteropathy, X-linkedâ€“like disorder caused by loss-of-function mutations in LRBA. Journal of Allergy and Clinical Immunology, 2015, 135, 217-227.e9.	2.9	223
75	Broad-spectrum antibodies against self-antigens and cytokines in RAG deficiency. Journal of Clinical Investigation, 2015, 125, 4135-4148.	8.2	159
76	Gene hunting in the genomic era: Approaches to diagnostic dilemmas in patients with primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2014, 134, 262-268.	2.9	34
77	A novel mutation in FOXN1 resulting in SCID: A case report and literature review. Clinical Immunology, 2014, 155, 30-32.	3.2	32
78	Leucine-rich repeat containing 8A (LRRC8A) is essential for T lymphocyte development and function. Journal of Experimental Medicine, 2014, 211, 929-942.	8.5	95
79	Lessons in gene hunting: AÂRAG1 mutation presenting with agammaglobulinemia and absence of B cells. Journal of Allergy and Clinical Immunology, 2014, 134, 983-985.e1.	2.9	22
80	Presence of hypogammaglobulinemia and abnormal antibody responses in GATA2 deficiency. Journal of Allergy and Clinical Immunology, 2014, 134, 223-226.	2.9	25
81	A novel disease-causing CD40L mutation reduces expression of CD40 ligand, but preserves CD40 binding capacity. Clinical Immunology, 2014, 153, 288-291.	3.2	9
82	Defective actin accumulation impairs human natural killer cell function in patients with dedicator of cytokinesis 8 deficiency. Journal of Allergy and Clinical Immunology, 2013, 131, 840-848.	2.9	113
83	A homozygous mucosa-associated lymphoid tissue 1 (MALT1) mutation in a family with combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2013, 132, 151-158.	2.9	124
84	Orbital Follicular Hyperplasia in Common Variable Immune Deficiency Syndrome. Ophthalmic Plastic and Reconstructive Surgery, 2013, 29, e160-e162.	0.8	1
85	Use of whole exome and genome sequencing in the identification of genetic causes of primary immunodeficiencies. Current Opinion in Allergy and Clinical Immunology, 2012, 12, 623-628.	2.3	65
86	A novel homozygous mutation in recombination activating gene 2 in 2 relatives with different clinical phenotypes: Omenn syndrome and hyper-IgM syndrome. Journal of Allergy and Clinical Immunology, 2012, 130, 1414-1416.	2.9	43
87	Predictors of clinical success in a multidisciplinary model of atopic dermatitis treatment. Allergy and Asthma Proceedings, 2011, 32, 377-383.	2.2	21
88	NOD2-associated diseases: Bridging innate immunity and autoinflammation. Clinical Immunology, 2010, 134, 251-261.	3.2	76
89	Leukocyte-versus microparticle-mediated tissue factor transfer during arteriolar thrombus development. Journal of Leukocyte Biology, 2005, 78, 1318-1326.	3.3	135
90	Hematopoietic cell-derived microparticle tissue factor contributes to fibrin formation during thrombus propagation. Blood, 2004, 104, 3190-3197.	1.4	323

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91	Accumulation of Tissue Factor into Developing Thrombi In Vivo Is Dependent upon Microparticle P-Selectin Glycoprotein Ligand 1 and Platelet P-Selectin. Journal of Experimental Medicine, 2003, 197, 1585-1598.	8.5	700
92	Filamin A, the Arp2/3 complex, and the morphology and function of cortical actin filaments in human melanoma cells. Journal of Cell Biology, 2001, 155, 511-518.	5.2	167
93	Dysregulation of the cGAS-STING Pathway in Monogenic Autoinflammation and Lupus. Frontiers in Immunology, 0, 13, .	4.8	10