

# Jennifer M Puck

## List of Publications by Year in descending order

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95  
papers

12,318  
citations

38738

50  
h-index

40976

93  
g-index

101  
all docs

101  
docs citations

101  
times ranked

9267  
citing authors

#	ARTICLE	IF	CITATIONS
1	Human Inborn Errors of Immunity: 2019 Update on the Classification from the International Union of Immunological Societies Expert Committee. <i>Journal of Clinical Immunology</i> , 2020, 40, 24-64.	3.8	881
2	International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. <i>Journal of Clinical Immunology</i> , 2018, 38, 96-128.	3.8	732
3	Primary Immunodeficiency Diseases: an Update on the Classification from the International Union of Immunological Societies Expert Committee for Primary Immunodeficiency 2015. <i>Journal of Clinical Immunology</i> , 2015, 35, 696-726.	3.8	621
4	Transplantation Outcomes for Severe Combined Immunodeficiency, 2000â€“2009. <i>New England Journal of Medicine</i> , 2014, 371, 434-446.	27.0	594
5	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
6	Human Inborn Errors of Immunity: 2019 Update of the IUIS Phenotypic Classification. <i>Journal of Clinical Immunology</i> , 2020, 40, 66-81.	3.8	525
7	Human severe combined immunodeficiency: Genetic, phenotypic, and functional diversity in one hundred eight infants. <i>Journal of Pediatrics</i> , 1997, 130, 378-387.	1.8	515
8	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. <i>Journal of Clinical Immunology</i> , 2018, 38, 129-143.	3.8	488
9	Primary Immunodeficiency Diseases: An Update on the Classification from the International Union of Immunological Societies Expert Committee for Primary Immunodeficiency. <i>Frontiers in Immunology</i> , 2014, 5, 162.	4.8	466
10	Human Inborn Errors of Immunity: 2022 Update on the Classification from the International Union of Immunological Societies Expert Committee. <i>Journal of Clinical Immunology</i> , 2022, 42, 1473-1507.	3.8	389
11	Development of population-based newborn screening for severe combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2005, 115, 391-398.	2.9	366
12	Hematopoietic stem cell transplantation for severe combined immunodeficiency in the neonatal period leads to superior thymic output and improved survival. <i>Blood</i> , 2002, 99, 872-878.	1.4	321
13	Establishing diagnostic criteria for severe combined immunodeficiency disease (SCID), leaky SCID, and Omenn syndrome: The Primary Immune Deficiency Treatment Consortium experience. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 1092-1098.	2.9	301
14	Journal Info Home About the Journal Editorial Board Archive Research Topics View Some Authors Review Guidelines Subscribe to Alerts Search Article Type Publication Date ..... Go Author Info Why Submit? Fees Article Types Author Guidelines Submission Checklist Contact Editorial Office Submit Manuscript Review ARTICLE Abstract Full Text PDF O Write a Comment Primary immunodeficiency diseases: an update on the classification from the International Union of Immunological Societies Expert Committee for Primary. <i>Frontiers in Immunology</i> , 2011, 2, 54.	4.8	294
15	Immune reconstitution and survival of 100 SCID patients postâ€“hematopoietic cell transplant: a PIDTC natural history study. <i>Blood</i> , 2017, 130, 2718-2727.	1.4	212
16	Lentiviral Gene Therapy Combined with Low-Dose Busulfan in Infants with SCID-X1. <i>New England Journal of Medicine</i> , 2019, 380, 1525-1534.	27.0	203
17	The 2015 IUIS Phenotypic Classification for Primary Immunodeficiencies. <i>Journal of Clinical Immunology</i> , 2015, 35, 727-738.	3.8	199
18	Polymer-stabilized Cas9 nanoparticles and modified repair templates increase genome editing efficiency. <i>Nature Biotechnology</i> , 2020, 38, 44-49.	17.5	198

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19	Newborn screening for severe combined immunodeficiency and T-cell lymphopenia in California: Results of the first 2 years. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 140-150.e7.	2.9	189
20	Newborn Sequencing in Genomic Medicine and Public Health. <i>Pediatrics</i> , 2017, 139, .	2.1	174
21	Severe combined immunodeficiencies and related disorders. <i>Nature Reviews Disease Primers</i> , 2015, 1, 15061.	30.5	173
22	Update on the safety and efficacy of retroviral gene therapy for immunodeficiency due to adenosine deaminase deficiency. <i>Blood</i> , 2016, 128, 45-54.	1.4	173
23	The Ever-Increasing Array of Novel Inborn Errors of Immunity: an Interim Update by the IUIS Committee. <i>Journal of Clinical Immunology</i> , 2021, 41, 666-679.	3.8	165
24	Laboratory technology for population-based screening for severe combined immunodeficiency in neonates: The winner is T-cell receptor excision circles. <i>Journal of Allergy and Clinical Immunology</i> , 2012, 129, 607-616.	2.9	161
25	Newborn Screening for Severe Combined Immunodeficiency and T-cell Lymphopenia in California, 2010â€“2017. <i>Pediatrics</i> , 2019, 143, .	2.1	148
26	A systematic analysis of recombination activity and genotype-phenotype correlation in human recombination-activating gene 1 deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 1099-1108.e12.	2.9	132
27	SCID genotype and 6-month posttransplant CD4 count predict survival and immune recovery. <i>Blood</i> , 2018, 132, 1737-1749.	1.4	128
28	Newborn screening for severe combined immunodeficiency and T cell lymphopenia. <i>Immunological Reviews</i> , 2019, 287, 241-252.	6.0	127
29	Population-based newborn screening for severe combined immunodeficiency: Steps toward implementation. <i>Journal of Allergy and Clinical Immunology</i> , 2007, 120, 760-768.	2.9	122
30	History and current status of newborn screening for severe combined immunodeficiency. <i>Seminars in Perinatology</i> , 2015, 39, 194-205.	2.5	116
31	The role of exome sequencing in newborn screening for inborn errors of metabolism. <i>Nature Medicine</i> , 2020, 26, 1392-1397.	30.7	112
32	Multisystem Anomalies in Severe Combined Immunodeficiency with Mutant <i>BCL11B</i> . <i>New England Journal of Medicine</i> , 2016, 375, 2165-2176.	27.0	104
33	Consensus approach for the management of severe combined immune deficiency caused by adenosine deaminase deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 852-863.	2.9	104
34	The Natural History of Children with Severe Combined Immunodeficiency: Baseline Features of the First Fifty Patients of the Primary Immune Deficiency Treatment Consortium Prospective Study 6901. <i>Journal of Clinical Immunology</i> , 2013, 33, 1156-1164.	3.8	100
35	A Markov model to analyze cost-effectiveness of screening for severe combined immunodeficiency (SCID). <i>Molecular Genetics and Metabolism</i> , 2011, 104, 383-389.	1.1	93
36	Newborn Screening for SCID Identifies Patients with Ataxia Telangiectasia. <i>Journal of Clinical Immunology</i> , 2013, 33, 540-549.	3.8	92

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37	An essential role for the Zn <sup>2+</sup> transporter ZIP7 in B cell development. <i>Nature Immunology</i> , 2019, 20, 350-361.	14.5	92
38	Excellent outcomes following hematopoietic cell transplantation for Wiskott-Aldrich syndrome: a PIDTC report. <i>Blood</i> , 2020, 135, 2094-2105.	1.4	87
39	A novel human autoimmune syndrome caused by combined hypomorphic and activating mutations in ZAP-70. <i>Journal of Experimental Medicine</i> , 2016, 213, 155-165.	8.5	83
40	<i>EXTL3</i> mutations cause skeletal dysplasia, immune deficiency, and developmental delay. <i>Journal of Experimental Medicine</i> , 2017, 214, 623-637.	8.5	76
41	Early vs. delayed diagnosis of severe combined immunodeficiency: A family perspective survey. <i>Clinical Immunology</i> , 2011, 138, 3-8.	3.2	73
42	Treatment of infants identified as having severe combined immunodeficiency by means of newborn screening. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 733-742.	2.9	73
43	Primary Immune Deficiency Treatment Consortium (PIDTC) report. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 335-347.e11.	2.9	65
44	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
45	The genetic landscape of severe combined immunodeficiency in the United States and Canada in the current era (2010-2018). <i>Journal of Allergy and Clinical Immunology</i> , 2019, 143, 405-407.	2.9	64
46	SCID newborn screening: What we've learned. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 417-426.	2.9	64
47	Abnormal B-cell maturation in the bone marrow of patients with germline mutations in PIK3CD. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 1032-1035.e6.	2.9	62
48	Deficient T Cell Receptor Excision Circles (TRECs) in autosomal recessive hyper IgE syndrome caused by DOCK8 mutation: Implications for pathogenesis and potential detection by newborn screening. <i>Clinical Immunology</i> , 2011, 141, 128-132.	3.2	57
49	Hematopoietic Cell Transplantation in Patients With Primary Immune Regulatory Disorders (PIRD): A Primary Immune Deficiency Treatment Consortium (PIDTC) Survey. <i>Frontiers in Immunology</i> , 2020, 11, 239.	4.8	57
50	Neonatal screening for severe combined immunodeficiency. <i>Current Opinion in Pediatrics</i> , 2011, 23, 667-673.	2.0	54
51	Diagnostic interpretation of genetic studies in patients with primary immunodeficiency diseases: A working group report of the Primary Immunodeficiency Diseases Committee of the American Academy of Allergy, Asthma & Immunology. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 145, 46-69.	2.9	54
52	Current Knowledge and Priorities for Future Research in Late Effects after Hematopoietic Stem Cell Transplantation (HCT) for Severe Combined Immunodeficiency Patients: A Consensus Statement from the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects after Pediatric HCT. <i>Biology of Blood and Marrow Transplantation</i> , 2017, 23, 379-387.	2.0	49
53	Successful newborn screening for SCID in the Navajo Nation. <i>Clinical Immunology</i> , 2015, 158, 29-34.	3.2	48
54	The case for newborn screening for severe combined immunodeficiency and related disorders. <i>Annals of the New York Academy of Sciences</i> , 2011, 1246, 108-117.	3.8	44

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55	Recommendations for Screening and Management of Late Effects in Patients with Severe Combined Immunodeficiency after Allogeneic Hematopoietic Cell Transplantation: A Consensus Statement from the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects after Pediatric HCT. <i>Biology of Blood and Marrow Transplantation</i> , 2017, 23, 1229-1240.	2.0	44
56	Lentivirus Mediated Correction of Artemis-Deficient Severe Combined Immunodeficiency. <i>Human Gene Therapy</i> , 2017, 28, 112-124.	2.7	44
57	Whole exome and whole genome sequencing with dried blood spot DNA without whole genome amplification. <i>Human Mutation</i> , 2018, 39, 167-171.	2.5	41
58	Lessons from the Wiskott-Aldrich Syndrome. <i>New England Journal of Medicine</i> , 2006, 355, 1759-1761.	27.0	39
59	Newborn Screening for Severe Combined Immunodeficiency in the US: Current Status and Approach to Management. <i>International Journal of Neonatal Screening</i> , 2017, 3, 15.	3.2	38
60	B-cell differentiation and IL-21 response in IL2RG/JAK3 SCID patients after hematopoietic stem cell transplantation. <i>Blood</i> , 2018, 131, 2967-2977.	1.4	37
61	Infections in Infants with SCID: Isolation, Infection Screening, and Prophylaxis in PIDTC Centers. <i>Journal of Clinical Immunology</i> , 2021, 41, 38-50.	3.8	36
62	Nijmegen Breakage Syndrome Detected by Newborn Screening for T Cell Receptor Excision Circles (TRECs). <i>Journal of Clinical Immunology</i> , 2015, 35, 227-233.	3.8	34
63	Primary Immune Deficiency Treatment Consortium (PIDTC) update. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 375-385.	2.9	33
64	Immunodeficiencies Associated with Abnormal Newborn Screening for T Cell and B Cell Lymphopenia. <i>Journal of Clinical Immunology</i> , 2017, 37, 363-374.	3.8	33
65	Reference intervals for lymphocyte subsets in preterm and term neonates without immune defects. <i>Journal of Allergy and Clinical Immunology</i> , 2019, 144, 1674-1683.	2.9	33
66	Mutations causing severe combined immunodeficiency: detection with a custom resequencing microarray. <i>Genetics in Medicine</i> , 2008, 10, 575-585.	2.4	31
67	Constrained chromatin accessibility in PU.1-mutated agammaglobulinemia patients. <i>Journal of Experimental Medicine</i> , 2021, 218, .	8.5	31
68	B-cell development and functions and therapeutic options in adenosine deaminase-deficient patients. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 799-806.e10.	2.9	30
69	Newborn Screening for Severe Combined Immunodeficiency in the United States. <i>Immunology and Allergy Clinics of North America</i> , 2019, 39, 1-11.	1.9	29
70	Neonatal screening for severe combined immune deficiency. <i>Current Opinion in Allergy and Clinical Immunology</i> , 2007, 7, 522-527.	2.3	28
71	Diagnostic assay to assist clinical decisions for unclassified severe combined immune deficiency. <i>Blood Advances</i> , 2020, 4, 2606-2610.	5.2	28
72	Outcomes following treatment for ADA-deficient severe combined immunodeficiency: a report from the PIDTC. <i>Blood</i> , 2022, 140, 685-705.	1.4	26

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73	Low Exposure Busulfan Conditioning to Achieve Sufficient Multilineage Chimerism in Patients with Severe Combined Immunodeficiency. <i>Biology of Blood and Marrow Transplantation</i> , 2019, 25, 1355-1362.	2.0	22
74	Adenosine Deaminase (ADA)â€“Deficient Severe Combined Immune Deficiency (SCID) in the US Immunodeficiency Network (USIDNet) Registry. <i>Journal of Clinical Immunology</i> , 2020, 40, 1124-1131.	3.8	19
75	Recommendations for uniform definitions used in newborn screening for severe combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 1428-1436.	2.9	19
76	Lentivector cryptic splicing mediates increase in CD34+ clones expressing truncated HMGA2 in human X-linked severe combined immunodeficiency. <i>Nature Communications</i> , 2022, 13, .	12.8	19
77	Neurologic eventâ€“free survival demonstrates a benefit for SCID patients diagnosed by newborn screening. <i>Blood Advances</i> , 2017, 1, 1694-1698.	5.2	14
78	When Screening for Severe Combined Immunodeficiency (SCID) with T Cell Receptor Excision Circles Is Not SCID: a Case-Based Review. <i>Journal of Clinical Immunology</i> , 2021, 41, 294-302.	3.8	13
79	Severe combined immunodeficiency: new advances in diagnosis and treatment. <i>Immunologic Research</i> , 2007, 38, 64-67.	2.9	12
80	Genomic Analysis of Historical Cases with Positive Newborn Screens for Short-Chain Acyl-CoA Dehydrogenase Deficiency Shows That a Validated Second-Tier Biochemical Test Can Replace Future Sequencing. <i>International Journal of Neonatal Screening</i> , 2020, 6, 41.	3.2	9
81	Poor T-cell receptor Î² repertoire diversity early posttransplant for severe combined immunodeficiency predicts failure of immune reconstitution. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 1113-1119.	2.9	8
82	Establishing Newborn Screening for SCID in the USA: Experience in California. <i>International Journal of Neonatal Screening</i> , 2021, 7, 72.	3.2	7
83	Inborn Errors of Immunity Associated With Type 2 Inflammation in the USIDNET Registry. <i>Frontiers in Immunology</i> , 2022, 13, 831279.	4.8	6
84	Longstanding Eosinophilia in a Case of Late Diagnosis Chronic Granulomatous Disease. <i>Journal of Clinical Immunology</i> , 2017, 37, 101-103.	3.8	4
85	Severe combined immunodeficiency in an infant with multiple congenital abnormalities. <i>Journal of Allergy and Clinical Immunology</i> , 1999, 103, 1222-1223.	2.9	3
86	Newborn Screening for Severe Combined Immunodeficiency. <i>Current Pediatrics Reports</i> , 2015, 3, 34-42.	4.0	3
87	Successful SCID gene therapy in infant with disseminated BCG. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 993-995.e1.	3.8	3
88	Granulocyte Transfusions in Patients with Chronic Granulomatous Disease Undergoing Hematopoietic Cell Transplantation or Gene Therapy. <i>Journal of Clinical Immunology</i> , 2022, 42, 1026-1035.	3.8	3
89	Unknown cytomegalovirus serostatus in primary immunodeficiency disorders: A new category of transplant recipients. <i>Transplant Infectious Disease</i> , 2021, 23, e13504.	1.7	2
90	USE OF GENOME DATA IN NEWBORNS AS A STARTING POINT FOR LIFE-LONG PRECISION MEDICINE. , 2016, , .		1

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91	Extended Follow-up After Hematopoietic Cell Transplantation for $\text{IL}12\beta$ Deficiency with Disseminated Mycobacterium avium Infection. Journal of Clinical Immunology, 2020, 40, 248-250.	3.8	1
92	Use of Rituximab for Refractory Immune Cytopenias Associated with Autoimmune Lymphoproliferative Syndrome (ALPS).. Blood, 2007, 110, 1319-1319.	1.4	1
93	Human Genomics in Immunology. , 2019, , 463-470.e1.		0
94	Expectations and experience: Parent and patient perspectives regarding treatment for Severe Combined Immunodeficiency (SCID). Clinical Immunology, 2021, 229, 108778.	3.2	0
95	Population-based newborn screening for severe combined immunodeficiency. Biology of Blood and Marrow Transplantation, 2008, 14, 78-80.	2.0	0