

# Jennifer M Puck

## List of Publications by Citations

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94  
papers

8,113  
citations

42  
h-index

90  
g-index

101  
ext. papers

10,709  
ext. citations

9.2  
avg, IF

6.07  
L-index

#	Paper	IF	Citations
94	International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. <i>Journal of Clinical Immunology</i> , <b>2018</b> , 38, 96-128	5.7	510
93	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> , <b>2020</b> , 40, 24-64	5.7	497
92	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> , <b>2015</b> , 35, 696-726	5.7	478
91	Human severe combined immunodeficiency: genetic, phenotypic, and functional diversity in one hundred eight infants. <i>Journal of Pediatrics</i> , <b>1997</b> , 130, 378-87	3.6	459
90	Transplantation outcomes for severe combined immunodeficiency, 2000-2009. <i>New England Journal of Medicine</i> , <b>2014</b> , 371, 434-46	59.2	457
89	Newborn screening for severe combined immunodeficiency in 11 screening programs in the United States. <i>JAMA - Journal of the American Medical Association</i> , <b>2014</b> , 312, 729-38	27.4	426
88	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. <i>Journal of Clinical Immunology</i> , <b>2018</b> , 38, 129-143	5.7	345
87	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> , <b>2014</b> , 5, 162	8.4	309
86	Development of population-based newborn screening for severe combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , <b>2005</b> , 115, 391-8	11.5	284
85	Hematopoietic stem cell transplantation for severe combined immunodeficiency in the neonatal period leads to superior thymic output and improved survival. <i>Blood</i> , <b>2002</b> , 99, 872-8	2.2	282
84	Human Inborn Errors of Immunity: 2019 Update of the IUIS Phenotypical Classification. <i>Journal of Clinical Immunology</i> , <b>2020</b> , 40, 66-81	5.7	267
83	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> , <b>2011</b> , 2, 54	8.4	266
82	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> , <b>2014</b> , 133, 1092-8	11.5	222
81	The 2015 IUIS Phenotypic Classification for Primary Immunodeficiencies. <i>Journal of Clinical Immunology</i> , <b>2015</b> , 35, 727-38	5.7	160
80	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> , <b>2013</b> , 132, 140-50	11.5	153
79	Update on the safety and efficacy of retroviral gene therapy for immunodeficiency due to adenosine deaminase deficiency. <i>Blood</i> , <b>2016</b> , 128, 45-54	2.2	133
78	Immune reconstitution and survival of 100 SCID patients post-hematopoietic cell transplant: a PIDTC natural history study. <i>Blood</i> , <b>2017</b> , 130, 2718-2727	2.2	129

77	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> , <b>2012</b> , 129, 607-16	11.5	123
76	Lentiviral Gene Therapy Combined with Low-Dose Busulfan in Infants with SCID-X1. <i>New England Journal of Medicine</i> , <b>2019</b> , 380, 1525-1534	59.2	122
75	Newborn Sequencing in Genomic Medicine and Public Health. <i>Pediatrics</i> , <b>2017</b> , 139,	7.4	109
74	Severe combined immunodeficiencies and related disorders. <i>Nature Reviews Disease Primers</i> , <b>2015</b> , 1, 15061	51.1	108
73	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> , <b>2014</b> , 133, 1099-1115	11.5	100
72	Population-based newborn screening for severe combined immunodeficiency: steps toward implementation. <i>Journal of Allergy and Clinical Immunology</i> , <b>2007</b> , 120, 760-8	11.5	97
71	SCID genotype and 6-month posttransplant CD4 count predict survival and immune recovery. <i>Blood</i> , <b>2018</b> , 132, 1737-1749	2.2	88
70	Newborn Screening for Severe Combined Immunodeficiency and T-cell Lymphopenia in California, 2010-2017. <i>Pediatrics</i> , <b>2019</b> , 143,	7.4	82
69	Polymer-stabilized Cas9 nanoparticles and modified repair templates increase genome editing efficiency. <i>Nature Biotechnology</i> , <b>2020</b> , 38, 44-49	44.5	79
68	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> , <b>2013</b> , 33, 1156-64	5.7	78
67	History and current status of newborn screening for severe combined immunodeficiency. <i>Seminars in Perinatology</i> , <b>2015</b> , 39, 194-205	3.3	77
66	A Markov model to analyze cost-effectiveness of screening for severe combined immunodeficiency (SCID). <i>Molecular Genetics and Metabolism</i> , <b>2011</b> , 104, 383-9	3.7	76
65	Consensus approach for the management of severe combined immune deficiency caused by adenosine deaminase deficiency. <i>Journal of Allergy and Clinical Immunology</i> , <b>2019</b> , 143, 852-863	11.5	71
64	Newborn screening for severe combined immunodeficiency and T-cell lymphopenia. <i>Immunological Reviews</i> , <b>2019</b> , 287, 241-252	11.3	70
63	Newborn screening for SCID identifies patients with ataxia telangiectasia. <i>Journal of Clinical Immunology</i> , <b>2013</b> , 33, 540-9	5.7	69
62	The Ever-Increasing Array of Novel Inborn Errors of Immunity: an Interim Update by the IUIS Committee. <i>Journal of Clinical Immunology</i> , <b>2021</b> , 41, 666-679	5.7	66
61	A novel human autoimmune syndrome caused by combined hypomorphic and activating mutations in ZAP-70. <i>Journal of Experimental Medicine</i> , <b>2016</b> , 213, 155-65	16.6	60
60	Early vs. delayed diagnosis of severe combined immunodeficiency: a family perspective survey. <i>Clinical Immunology</i> , <b>2011</b> , 138, 3-8	9	60

59	Multisystem Anomalies in Severe Combined Immunodeficiency with Mutant BCL11B. <i>New England Journal of Medicine</i> , <b>2016</b> , 375, 2165-2176	59.2	57
58	mutations cause skeletal dysplasia, immune deficiency, and developmental delay. <i>Journal of Experimental Medicine</i> , <b>2017</b> , 214, 623-637	16.6	54
57	An essential role for the Zn transporter ZIP7 in B cell development. <i>Nature Immunology</i> , <b>2019</b> , 20, 350-361	19.1	54
56	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> , <b>2011</b> , 141, 128-32	9	50
55	Treatment of infants identified as having severe combined immunodeficiency by means of newborn screening. <i>Journal of Allergy and Clinical Immunology</i> , <b>2017</b> , 139, 733-742	11.5	49
54	Excellent outcomes following hematopoietic cell transplantation for Wiskott-Aldrich syndrome: a PIDTC report. <i>Blood</i> , <b>2020</b> , 135, 2094-2105	2.2	46
53	Primary Immune Deficiency Treatment Consortium (PIDTC) report. <i>Journal of Allergy and Clinical Immunology</i> , <b>2014</b> , 133, 335-47	11.5	42
52	Abnormal B-cell maturation in the bone marrow of patients with germline mutations in PIK3CD. <i>Journal of Allergy and Clinical Immunology</i> , <b>2017</b> , 139, 1032-1035.e6	11.5	42
51	Neonatal screening for severe combined immunodeficiency. <i>Current Opinion in Pediatrics</i> , <b>2011</b> , 23, 667-33	33	42
50	Outcomes and Treatment Strategies for Autoimmunity and Hyperinflammation in Patients with RAG Deficiency. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , <b>2019</b> , 7, 1970-1985.e4	5.4	41
49	The role of exome sequencing in newborn screening for inborn errors of metabolism. <i>Nature Medicine</i> , <b>2020</b> , 26, 1392-1397	50.5	41
48	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> , <b>2019</b> , 143, 405-407	11.5	35
47	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> , <b>2017</b> , 23, 379-387	4.7	34
46	Lessons from the Wiskott-Aldrich syndrome. <i>New England Journal of Medicine</i> , <b>2006</b> , 355, 1759-61	59.2	34
45	Lentivirus Mediated Correction of Artemis-Deficient Severe Combined Immunodeficiency. <i>Human Gene Therapy</i> , <b>2017</b> , 28, 112-124	4.8	32
44	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> , <b>2020</b> , 145, 46-69	11.5	32
43	The case for newborn screening for severe combined immunodeficiency and related disorders. <i>Annals of the New York Academy of Sciences</i> , <b>2011</b> , 1246, 108-17	6.5	29
42	Successful newborn screening for SCID in the Navajo Nation. <i>Clinical Immunology</i> , <b>2015</b> , 158, 29-34	9	28

41	Mutations causing severe combined immunodeficiency: detection with a custom resequencing microarray. <i>Genetics in Medicine</i> , <b>2008</b> , 10, 575-85	8.1	28
40	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> , <b>2017</b> ,	4.7	27
39	B-cell development and functions and therapeutic options in adenosine deaminase-deficient patients. <i>Journal of Allergy and Clinical Immunology</i> , <b>2014</b> , 133, 799-806.e10	11.5	27
38	Neonatal screening for severe combined immune deficiency. <i>Current Opinion in Allergy and Clinical Immunology</i> , <b>2007</b> , 7, 522-7	3.3	26
37	Hematopoietic Cell Transplantation in Patients With Primary Immune Regulatory Disorders (PIRD): A Primary Immune Deficiency Treatment Consortium (PIDTC) Survey. <i>Frontiers in Immunology</i> , <b>2020</b> , 11, 239	8.4	25
36	Whole exome and whole genome sequencing with dried blood spot DNA without whole genome amplification. <i>Human Mutation</i> , <b>2018</b> , 39, 167-171	4.7	23
35	Reference intervals for lymphocyte subsets in preterm and term neonates without immune defects. <i>Journal of Allergy and Clinical Immunology</i> , <b>2019</b> , 144, 1674-1683	11.5	23
34	Primary Immune Deficiency Treatment Consortium (PIDTC) update. <i>Journal of Allergy and Clinical Immunology</i> , <b>2016</b> , 138, 375-85	11.5	22
33	B-cell differentiation and IL-21 response in SCID patients after hematopoietic stem cell transplantation. <i>Blood</i> , <b>2018</b> , 131, 2967-2977	2.2	22
32	Immunodeficiencies Associated with Abnormal Newborn Screening for T Cell and B Cell Lymphopenia. <i>Journal of Clinical Immunology</i> , <b>2017</b> , 37, 363-374	5.7	21
31	Nijmegen breakage syndrome detected by newborn screening for T cell receptor excision circles (TRECs). <i>Journal of Clinical Immunology</i> , <b>2015</b> , 35, 227-33	5.7	21
30	Newborn Screening for Severe Combined Immunodeficiency in the US: Current Status and Approach to Management. <i>International Journal of Neonatal Screening</i> , <b>2017</b> , 3,	2.6	18
29	Newborn Screening for Severe Combined Immunodeficiency in the United States: Lessons Learned. <i>Immunology and Allergy Clinics of North America</i> , <b>2019</b> , 39, 1-11	3.3	18
28	Diagnostic assay to assist clinical decisions for unclassified severe combined immune deficiency. <i>Blood Advances</i> , <b>2020</b> , 4, 2606-2610	7.8	14
27	SCID newborn screening: What we've learned. <i>Journal of Allergy and Clinical Immunology</i> , <b>2021</b> , 147, 417-425	11.5	14
26	Low Exposure Busulfan Conditioning to Achieve Sufficient Multilineage Chimerism in Patients with Severe Combined Immunodeficiency. <i>Biology of Blood and Marrow Transplantation</i> , <b>2019</b> , 25, 1355-1362	4.7	11
25	Severe combined immunodeficiency: new advances in diagnosis and treatment. <i>Immunologic Research</i> , <b>2007</b> , 38, 64-7	4.3	11
24	Neurologic event-free survival demonstrates a benefit for SCID patients diagnosed by newborn screening. <i>Blood Advances</i> , <b>2017</b> , 1, 1694-1698	7.8	10

23	Adenosine Deaminase (ADA)-Deficient Severe Combined Immune Deficiency (SCID) in the US Immunodeficiency Network (USIDNet) Registry. <i>Journal of Clinical Immunology</i> , <b>2020</b> , 40, 1124-1131	5.7	6
22	Constrained chromatin accessibility in PU.1-mutated agammaglobulinemia patients. <i>Journal of Experimental Medicine</i> , <b>2021</b> , 218,	16.6	6
21	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> , <b>2021</b> , 41, 294-302	5.7	6
20	Infections in Infants with SCID: Isolation, Infection Screening, and Prophylaxis in PIDTC Centers. <i>Journal of Clinical Immunology</i> , <b>2021</b> , 41, 38-50	5.7	5
19	Longstanding Eosinophilia in a Case of Late Diagnosis Chronic Granulomatous Disease. <i>Journal of Clinical Immunology</i> , <b>2017</b> , 37, 101-103	5.7	3
18	Severe combined immunodeficiency in an infant with multiple congenital abnormalities. <i>Journal of Allergy and Clinical Immunology</i> , <b>1999</b> , 103, 1222-3	11.5	3
17	Recommendations for uniform definitions used in newborn screening for severe combined immunodeficiency. <i>Journal of Allergy and Clinical Immunology</i> , <b>2021</b> ,	11.5	3
16	Newborn Screening for Severe Combined Immunodeficiency. <i>Current Pediatrics Reports</i> , <b>2015</b> , 3, 34-42	0.7	2
15	DNA from dried blood spots yields high quality sequences for exome analysis		2
14	Perturbation robustness analyses reveal important parameters in variant interpretation pipelines		2
13	Poor T-cell receptor repertoire diversity early posttransplant for severe combined immunodeficiency predicts failure of immune reconstitution. <i>Journal of Allergy and Clinical Immunology</i> , <b>2021</b> ,	11.5	2
12	Inborn Errors of Immunity Associated With Type 2 Inflammation in the USIDNET Registry.. <i>Frontiers in Immunology</i> , <b>2022</b> , 13, 831279	8.4	2
11	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> , <b>2020</b> , 6,	2.6	1
10	Use of Rituximab for Refractory Immune Cytopenias Associated with Autoimmune Lymphoproliferative Syndrome (ALPS).. <i>Blood</i> , <b>2007</b> , 110, 1319-1319	2.2	1
9	Extended Follow-up After Hematopoietic Cell Transplantation for B220 Deficiency with Disseminated Mycobacterium avium Infection. <i>Journal of Clinical Immunology</i> , <b>2020</b> , 40, 248-250	5.7	1
8	Successful SCID gene therapy in infant with disseminated BCG. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , <b>2021</b> , 9, 993-995.e1	5.4	1
7	Unknown cytomegalovirus serostatus in primary immunodeficiency disorders: A new category of transplant recipients. <i>Transplant Infectious Disease</i> , <b>2021</b> , 23, e13504	2.7	0
6	Granulocyte Transfusions in Patients with Chronic Granulomatous Disease Undergoing Hematopoietic Cell Transplantation or Gene Therapy.. <i>Journal of Clinical Immunology</i> , <b>2022</b> , 1	5.7	0

5 Prenatal Diagnosis of Primary Immunodeficiency Diseases 628-645

4 Prenatal Diagnosis of Primary Immunodeficiency Diseases **2021**, 982-1001

3 Human Genomics in Immunology **2019**, 463-470.e1

2 Expectations and experience: Parent and patient perspectives regarding treatment for Severe Combined Immunodeficiency (SCID). *Clinical Immunology*, **2021**, 229, 108778 9

1 Population-based newborn screening for severe combined immunodeficiency. *Biology of Blood and Marrow Transplantation*, **2008**, 14, 78-80 4-7