Jennifer M Puck

List of Publications by Year in Descending Order

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	Inborn Errors of Immunity Associated With Type 2 Inflammation in the USIDNET Registry.. *Frontiers in Immunology*, **2022**, 13, 831279	8.4	2
93	Granulocyte Transfusions in Patients with Chronic Granulomatous Disease Undergoing Hematopoietic Cell Transplantation or Gene Therapy.. *Journal of Clinical Immunology*, **2022**, 1	5.7	0
92	Prenatal Diagnosis of Primary Immunodeficiency Diseases **2021**, 982-1001		
91	Constrained chromatin accessibility in PU.1-mutated agammaglobulinemia patients. *Journal of Experimental Medicine*, **2021**, 218,	16.6	6
90	Unknown cytomegalovirus serostatus in primary immunodeficiency disorders: A new category of transplant recipients. *Transplant Infectious Disease*, **2021**, 23, e13504	2.7	0
89	Infections in Infants with SCID: Isolation, Infection Screening, and Prophylaxis in PIDTC Centers. *Journal of Clinical Immunology*, **2021**, 41, 38-50	5.7	5
88	Successful SCID gene therapy in infant with disseminated BCG. *Journal of Allergy and Clinical Immunology: in Practice*, **2021**, 9, 993-995.e1	5.4	1
87	When Screening for Severe Combined Immunodeficiency (SCID) with T Cell Receptor Excision Circles Is Not SCID: a Case-Based Review. *Journal of Clinical Immunology*, **2021**, 41, 294-302	5.7	6
86	The Ever-Increasing Array of Novel Inborn Errors of Immunity: an Interim Update by the IUIS Committee. *Journal of Clinical Immunology*, **2021**, 41, 666-679	5.7	66
85	SCID newborn screening: What we've learned. *Journal of Allergy and Clinical Immunology*, **2021**, 147, 417-426	11.5	14
84	Expectations and experience: Parent and patient perspectives regarding treatment for Severe Combined Immunodeficiency (SCID). *Clinical Immunology*, **2021**, 229, 108778	9	
83	Poor T-cell receptor repertoire diversity early posttransplant for severe combined immunodeficiency predicts failure of immune reconstitution. *Journal of Allergy and Clinical Immunology*, **2021**,	11.5	2
82	Recommendations for uniform definitions used in newborn screening for severe combined immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2021**,	11.5	3
81	Excellent outcomes following hematopoietic cell transplantation for Wiskott-Aldrich syndrome: a PIDTC report. *Blood*, **2020**, 135, 2094-2105	2.2	46
80	Diagnostic assay to assist clinical decisions for unclassified severe combined immune deficiency. *Blood Advances*, **2020**, 4, 2606-2610	7.8	14
79	Hematopoietic Cell Transplantation in Patients With Primary Immune Regulatory Disorders (PIRD): A Primary Immune Deficiency Treatment Consortium (PIDTC) Survey. *Frontiers in Immunology*, **2020**, 11, 239	8.4	25
78	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. *International Journal of Neonatal Screening*, **2020**, 6,	2.6	1

77	Human Inborn Errors of Immunity: 2019 Update on the Classification from the International Union of Immunological Societies Expert Committee. *Journal of Clinical Immunology*, **2020**, 40, 24-64	5.7	497
76	Human Inborn Errors of Immunity: 2019 Update of the IUIS Phenotypical Classification. *Journal of Clinical Immunology*, **2020**, 40, 66-81	5.7	267
75	Extended Follow-up After Hematopoietic Cell Transplantation for IB Deficiency with Disseminated Mycobacterium avium Infection. *Journal of Clinical Immunology*, **2020**, 40, 248-250	5.7	1
74	Polymer-stabilized Cas9 nanoparticles and modified repair templates increase genome editing efficiency. *Nature Biotechnology*, **2020**, 38, 44-49	44.5	79
73	The role of exome sequencing in newborn screening for inborn errors of metabolism. *Nature Medicine*, **2020**, 26, 1392-1397	50.5	41
72	Adenosine Deaminase (ADA)-Deficient Severe Combined Immune Deficiency (SCID) in the US Immunodeficiency Network (USIDNet) Registry. *Journal of Clinical Immunology*, **2020**, 40, 1124-1131	5.7	6
71	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. *Journal of Allergy and Clinical Immunology*, **2020**, 145, 46-69	11.5	32
70	Newborn Screening for Severe Combined Immunodeficiency and T-cell Lymphopenia in California, 2010-2017. *Pediatrics*, **2019**, 143,	7.4	82
69	An essential role for the Zn transporter ZIP7 in B cell development. *Nature Immunology*, **2019**, 20, 350-361	19.1	54
68	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	5.4	41
67	Lentiviral Gene Therapy Combined with Low-Dose Busulfan in Infants with SCID-X1. *New England Journal of Medicine*, **2019**, 380, 1525-1534	59.2	122
66	Low Exposure Busulfan Conditioning to Achieve Sufficient Multilineage Chimerism in Patients with Severe Combined Immunodeficiency. *Biology of Blood and Marrow Transplantation*, **2019**, 25, 1355-1362	4.7	11
65	Reference intervals for lymphocyte subsets in preterm and term neonates without immune defects. *Journal of Allergy and Clinical Immunology*, **2019**, 144, 1674-1683	11.5	23
64	Human Genomics in Immunology **2019**, 463-470.e1		
63	Newborn screening for severe combined immunodeficiency and T-cell lymphopenia. *Immunological Reviews*, **2019**, 287, 241-252	11.3	70
62	Newborn Screening for Severe Combined Immunodeficiency in the United States: Lessons Learned. *Immunology and Allergy Clinics of North America*, **2019**, 39, 1-11	3.3	18
61	Consensus approach for the management of severe combined immune deficiency caused by adenosine deaminase deficiency. *Journal of Allergy and Clinical Immunology*, **2019**, 143, 852-863	11.5	71
60	The genetic landscape of severe combined immunodeficiency in the United States and Canada in the current era (2010-2018). *Journal of Allergy and Clinical Immunology*, **2019**, 143, 405-407	11.5	35

59	Whole exome and whole genome sequencing with dried blood spot DNA without whole genome amplification. *Human Mutation*, **2018**, 39, 167-171	4.7	23
58	International Union of Immunological Societies: 2017 Primary Immunodeficiency Diseases Committee Report on Inborn Errors of Immunity. *Journal of Clinical Immunology*, **2018**, 38, 96-128	5.7	510
57	The 2017 IUIS Phenotypic Classification for Primary Immunodeficiencies. *Journal of Clinical Immunology*, **2018**, 38, 129-143	5.7	345
56	SCID genotype and 6-month posttransplant CD4 count predict survival and immune recovery. *Blood* , **2018**, 132, 1737-1749	2.2	88
55	B-cell differentiation and IL-21 response in SCID patients after hematopoietic stem cell transplantation. *Blood*, **2018**, 131, 2967-2977	2.2	22
54	Newborn Sequencing in Genomic Medicine and Public Health. *Pediatrics*, **2017**, 139,	7.4	109
53	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. *Biology of Blood and Marrow Transplantation*, **2017**,	4.7	34
52	mutations cause skeletal dysplasia, immune deficiency, and developmental delay. *Journal of Experimental Medicine*, **2017**, 214, 623-637	16.6	54
51	Treatment of infants identified as having severe combined immunodeficiency by means of newborn screening. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 733-742	11.5	49
50	Recommendations for Screening and Management of Late Effects in Patients with Severe Combined Immunodeficiency after Allogenic Hematopoietic Cell Transplantation: A Consensus Statement from the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects after Pediatric HCT. *Biology of Blood and Marrow Transplantation*, **2017**,	4.7	27
49	Immunodeficiencies Associated with Abnormal Newborn Screening for T Cell and B Cell Lymphopenia. *Journal of Clinical Immunology*, **2017**, 37, 363-374	5.7	21
48	Longstanding Eosinophilia in a Case of Late Diagnosis Chronic Granulomatous Disease. *Journal of Clinical Immunology*, **2017**, 37, 101-103	5.7	3
47	Immune reconstitution and survival of 100 SCID patients post-hematopoietic cell transplant: a PIDTC natural history study. *Blood*, **2017**, 130, 2718-2727	2.2	129
46	Lentivirus Mediated Correction of Artemis-Deficient Severe Combined Immunodeficiency. *Human Gene Therapy*, **2017**, 28, 112-124	4.8	32
45	Abnormal B-cell maturation in the bone marrow of patients with germline mutations in PIK3CD. *Journal of Allergy and Clinical Immunology*, **2017**, 139, 1032-1035.e6	11.5	42
44	Neurologic event-free survival demonstrates a benefit for SCID patients diagnosed by newborn screening. *Blood Advances*, **2017**, 1, 1694-1698	7.8	10
43	Newborn Screening for Severe Combined Immunodeficiency in the US: Current Status and Approach to Management. *International Journal of Neonatal Screening*, **2017**, 3,	2.6	18
42	A novel human autoimmune syndrome caused by combined hypomorphic and activating mutations in ZAP-70. *Journal of Experimental Medicine*, **2016**, 213, 155-65	16.6	60

41	Multisystem Anomalies in Severe Combined Immunodeficiency with Mutant BCL11B. *New England Journal of Medicine*, **2016**, 375, 2165-2176	59.2	57
40	Primary Immune Deficiency Treatment Consortium (PIDTC) update. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 375-85	11.5	22
39	Update on the safety and efficacy of retroviral gene therapy for immunodeficiency due to adenosine deaminase deficiency. *Blood*, **2016**, 128, 45-54	2.2	133
38	Newborn Screening for Severe Combined Immunodeficiency. *Current Pediatrics Reports*, **2015**, 3, 34-42	0.7	2
37	History and current status of newborn screening for severe combined immunodeficiency. *Seminars in Perinatology*, **2015**, 39, 194-205	3.3	77
36	Primary Immunodeficiency Diseases: an Update on the Classification from the International Union of Immunological Societies Expert Committee for Primary Immunodeficiency 2015. *Journal of Clinical Immunology*, **2015**, 35, 696-726	5.7	478
35	The 2015 IUIS Phenotypic Classification for Primary Immunodeficiencies. *Journal of Clinical Immunology*, **2015**, 35, 727-38	5.7	160
34	Severe combined immunodeficiencies and related disorders. *Nature Reviews Disease Primers*, **2015**, 1, 15061	51.1	108
33	Successful newborn screening for SCID in the Navajo Nation. *Clinical Immunology*, **2015**, 158, 29-34	9	28
32	Nijmegen breakage syndrome detected by newborn screening for T cell receptor excision circles (TRECs). *Journal of Clinical Immunology*, **2015**, 35, 227-33	5.7	21
31	B-cell development and functions and therapeutic options in adenosine deaminase-deficient patients. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 799-806.e10	11.5	27
30	Establishing diagnostic criteria for severe combined immunodeficiency disease (SCID), leaky SCID, and Omenn syndrome: the Primary Immune Deficiency Treatment Consortium experience. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 1092-8	11.5	222
29	A systematic analysis of recombination activity and genotype-phenotype correlation in human recombination-activating gene 1 deficiency. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 1099-108	11.5	100
28	Primary Immune Deficiency Treatment Consortium (PIDTC) report. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 335-47	11.5	42
27	Transplantation outcomes for severe combined immunodeficiency, 2000-2009. *New England Journal of Medicine*, **2014**, 371, 434-46	59.2	457
26	Primary immunodeficiency diseases: an update on the classification from the international union of immunological societies expert committee for primary immunodeficiency. *Frontiers in Immunology*, **2014**, 5, 162	8.4	309
25	Newborn screening for severe combined immunodeficiency in 11 screening programs in the United States. *JAMA - Journal of the American Medical Association*, **2014**, 312, 729-38	27.4	426
24	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. *Journal of Clinical Immunology*, **2013**, 33, 1156-64	5.7	78

23	Newborn screening for SCID identifies patients with ataxia telangiectasia. *Journal of Clinical Immunology*, **2013**, 33, 540-9	5.7	69
22	Newborn screening for severe combined immunodeficiency and T-cell lymphopenia in California: results of the first 2 years. *Journal of Allergy and Clinical Immunology*, **2013**, 132, 140-50	11.5	153
21	Laboratory technology for population-based screening for severe combined immunodeficiency in neonates: the winner is T-cell receptor excision circles. *Journal of Allergy and Clinical Immunology*, **2012**, 129, 607-16	11.5	123
20	A Markov model to analyze cost-effectiveness of screening for severe combined immunodeficiency (SCID). *Molecular Genetics and Metabolism*, **2011**, 104, 383-9	3.7	76
19	The case for newborn screening for severe combined immunodeficiency and related disorders. *Annals of the New York Academy of Sciences*, **2011**, 1246, 108-17	6.5	29
18	Early vs. delayed diagnosis of severe combined immunodeficiency: a family perspective survey. *Clinical Immunology*, **2011**, 138, 3-8	9	60
17	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. *Clinical Immunology*, **2011**, 141, 128-32	9	50
16	Primary immunodeficiency diseases: an update on the classification from the international union of immunological societies expert committee for primary immunodeficiency. *Frontiers in Immunology*, **2011**, 2, 54	8.4	266
15	Neonatal screening for severe combined immunodeficiency. *Current Opinion in Pediatrics*, **2011**, 23, 667-73	3.2	42
14	Mutations causing severe combined immunodeficiency: detection with a custom resequencing microarray. *Genetics in Medicine*, **2008**, 10, 575-85	8.1	28
13	Population-based newborn screening for severe combined immunodeficiency. *Biology of Blood and Marrow Transplantation*, **2008**, 14, 78-80	4.7	
12	Population-based newborn screening for severe combined immunodeficiency: steps toward implementation. *Journal of Allergy and Clinical Immunology*, **2007**, 120, 760-8	11.5	97
11	Severe combined immunodeficiency: new advances in diagnosis and treatment. *Immunologic Research*, **2007**, 38, 64-7	4.3	11
10	Neonatal screening for severe combined immune deficiency. *Current Opinion in Allergy and Clinical Immunology*, **2007**, 7, 522-7	3.3	26
9	Use of Rituximab for Refractory Immune Cytopenias Associated with Autoimmune Lymphoproliferative Syndrome (ALPS).. *Blood*, **2007**, 110, 1319-1319	2.2	1
8	Lessons from the Wiskott-Aldrich syndrome. *New England Journal of Medicine*, **2006**, 355, 1759-61	59.2	34
7	Development of population-based newborn screening for severe combined immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2005**, 115, 391-8	11.5	284
6	Hematopoietic stem cell transplantation for severe combined immunodeficiency in the neonatal period leads to superior thymic output and improved survival. *Blood*, **2002**, 99, 872-8	2.2	282

5	Severe combined immunodeficiency in an infant with multiple congenital abnormalities. *Journal of Allergy and Clinical Immunology*, **1999**, 103, 1222-3	11.5	3
4	Human severe combined immunodeficiency: genetic, phenotypic, and functional diversity in one hundred eight infants. *Journal of Pediatrics*, **1997**, 130, 378-87	3.6	459
3	DNA from dried blood spots yields high quality sequences for exome analysis		2
2	Perturbation robustness analyses reveal important parameters in variant interpretation pipelines		2
1	Prenatal Diagnosis of Primary Immunodeficiency Diseases628-645		