

# Jennifer A Wambach

## List of Publications by Year in descending order

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62  
papers

1,816  
citations

430442

18  
h-index

288905

40  
g-index

66  
all docs

66  
docs citations

66  
times ranked

2632  
citing authors

#	ARTICLE	IF	CITATIONS
1	Differentiation of Human Pluripotent Stem Cells into Functional Lung Alveolar Epithelial Cells. <i>Cell Stem Cell</i> , 2017, 21, 472-488.e10.	5.2	406
2	Genotype-Phenotype Correlations for Infants and Children with ABCA3 Deficiency. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2014, 189, 1538-1543.	2.5	168
3	Novel <i>FOXF1</i> Mutations in Sporadic and Familial Cases of Alveolar Capillary Dysplasia with Misaligned Pulmonary Veins Imply a Role for its DNA Binding Domain. <i>Human Mutation</i> , 2013, 34, 801-811.	1.1	97
4	Population and Disease-Based Prevalence of the Common Mutations Associated With Surfactant Deficiency. <i>Pediatric Research</i> , 2008, 63, 645-649.	1.1	94
5	Single <i>ABCA3</i> Mutations Increase Risk for Neonatal Respiratory Distress Syndrome. <i>Pediatrics</i> , 2012, 130, e1575-e1582.	1.0	93
6	Women's Lifelong Exposure to Neighborhood Poverty and Low Birth Weight: A Population-Based Study. <i>Maternal and Child Health Journal</i> , 2009, 13, 326-33.	0.7	75
7	Outcomes of Lung Transplantation for Infants and Children with Genetic Disorders of Surfactant Metabolism. <i>Journal of Pediatrics</i> , 2017, 184, 157-164.e2.	0.9	66
8	Lysosomal Storage and Albinism Due to Effects of a De Novo <i>CLCN7</i> Variant on Lysosomal Acidification. <i>American Journal of Human Genetics</i> , 2019, 104, 1127-1138.	2.6	59
9	Gastrointestinal polyps in children: Advances in molecular genetics, diagnosis, and management. <i>Journal of Pediatrics</i> , 2001, 138, 621-628.	0.9	55
10	Infants with Atypical Presentations of Alveolar Capillary Dysplasia with Misalignment of the Pulmonary Veins Who Underwent Bilateral Lung Transplantation. <i>Journal of Pediatrics</i> , 2018, 194, 158-164.e1.	0.9	48
11	An intronic <i>ABCA3</i> mutation that is responsible for respiratory disease. <i>Pediatric Research</i> , 2012, 71, 633-637.	1.1	46
12	Two deletions overlapping a distant <i>FOXF1</i> enhancer unravel the role of lncRNA <i>LINC01081</i> in etiology of alveolar capillary dysplasia with misalignment of pulmonary veins. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2013-2019.	0.7	46
13	Functional Characterization of <i>ATP-Binding Cassette Transporter A3</i> Mutations from Infants with Respiratory Distress Syndrome. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2016, 55, 716-721.	1.4	44
14	Bi-allelic <i>POLR3A</i> Loss-of-Function Variants Cause Autosomal-Recessive Wiedemann-Rautenstrauch Syndrome. <i>American Journal of Human Genetics</i> , 2018, 103, 968-975.	2.6	43
15	Successful sulfonylurea treatment of an insulin-naïve neonate with diabetes mellitus due to a <i>KCNJ11</i> mutation. <i>Pediatric Diabetes</i> , 2009, 11, 286-288.	1.2	35
16	Surfactant Protein-C Promoter Variants Associated With Neonatal Respiratory Distress Syndrome Reduce Transcription. <i>Pediatric Research</i> , 2010, 68, 216-220.	1.1	34
17	Prevention and Treatment of Respiratory Distress Syndrome in Preterm Neonates. <i>Neonatal Network: NN</i> , 2018, 37, 169-177.	0.1	32
18	Phenotypic expansion of <i>KMT2D</i> -related disorder: Beyond Kabuki syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1053-1065.	0.7	23

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19	Neuroendocrine Cell Hyperplasia of Infancy. Clinical Score and Comorbidities. <i>Annals of the American Thoracic Society</i> , 2020, 17, 724-728.	1.5	21
20	Survival among children with "Lethal" congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene ( <i>GLDN</i> ). <i>Human Mutation</i> , 2017, 38, 1477-1484.	1.1	19
21	Heterozygous variants in <i>MYBPC1</i> are associated with an expanded neuromuscular phenotype beyond arthrogryposis. <i>Human Mutation</i> , 2019, 40, 1115-1126.	1.1	19
22	Functional Genomics of <i>ABCA3</i> Variants. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2020, 63, 436-443.	1.4	19
23	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. <i>Clinical Epigenetics</i> , 2019, 11, 60.	1.8	18
24	De novo 9q gain in an infant with tetralogy of Fallot with absent pulmonary valve: Patient report and review of congenital heart disease in 9q duplication syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2966-2974.	0.7	16
25	The common <i>ABCA3</i> <sup>E292V</sup> variant disrupts AT2 cell quality control and increases susceptibility to lung injury and aberrant remodeling. <i>American Journal of Physiology - Lung Cellular and Molecular Physiology</i> , 2021, 321, L291-L307.	1.3	16
26	Remission of Severe Neonatal Diabetes With Very Early Sulfonylurea Treatment. <i>Diabetes Care</i> , 2015, 38, e38-e39.	4.3	15
27	Genetic Factors Contribute to Risk for Neonatal Respiratory Distress Syndrome among Moderately Preterm, Late Preterm, and Term Infants. <i>Journal of Pediatrics</i> , 2016, 172, 69-74.e2.	0.9	14
28	LINE- and <i>Alu</i> -containing genomic instability hotspot at 16q24.1 associated with recurrent and nonrecurrent CNV deletions causative for ACDMPV. <i>Human Mutation</i> , 2018, 39, 1916-1925.	1.1	14
29	Functional characterization of four ATP-binding cassette transporter A3 gene ( <i>ABCA3</i> ) variants. <i>Human Mutation</i> , 2020, 41, 1298-1307.	1.1	13
30	Gene Therapy Potential for Genetic Disorders of Surfactant Dysfunction. <i>Frontiers in Genome Editing</i> , 2021, 3, 785829.	2.7	13
31	Recombination as a mechanism for sporadic mutation in the surfactant protein-C gene. <i>Pediatric Pulmonology</i> , 2008, 43, 443-450.	1.0	11
32	Functional characterization of biallelic <i>RTTN</i> variants identified in an infant with microcephaly, simplified gyral pattern, pontocerebellar hypoplasia, and seizures. <i>Pediatric Research</i> , 2018, 84, 435-441.	1.1	11
33	CemOrange2 fusions facilitate multifluorophore subcellular imaging in <i>C. elegans</i> . <i>PLoS ONE</i> , 2019, 14, e0214257.	1.1	11
34	Discovery of a novel <i>CHD7</i> CHARGE syndrome variant by integrated omics analyses. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 544-548.	0.7	10
35	New clinical practice guidelines on the classification, evaluation and management of childhood interstitial lung disease in infants: what do they mean?. <i>Expert Review of Respiratory Medicine</i> , 2014, 8, 653-655.	1.0	9
36	Accelerating Scientific Advancement for Pediatric Rare Lung Disease Research. Report from a National Institutes of Health "NHLBI Workshop, September 3 and 4, 2015. <i>Annals of the American Thoracic Society</i> , 2016, 13, 385-393.	1.5	9

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37	Phenotype and response to growth hormone therapy in siblings with B4GALT7 deficiency. <i>Bone</i> , 2019, 124, 14-21.	1.4	9
38	A dominant negative variant of <i>RAB5B</i> disrupts maturation of surfactant protein B and surfactant protein C. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	9
39	Neonatal Outcomes Differ after Spontaneous and Indicated Preterm Birth. <i>American Journal of Perinatology</i> , 2018, 35, 494-502.	0.6	8
40	Childhood rare lung disease in the 21st century: omics technology advances accelerating discovery. <i>Pediatric Pulmonology</i> , 2020, 55, 1828-1837.	1.0	8
41	Synonymous <i>ABCA3</i> Variants Do Not Increase Risk for Neonatal Respiratory Distress Syndrome. <i>Journal of Pediatrics</i> , 2014, 164, 1316-1321.e3.	0.9	6
42	Magnetic Resonance Imaging characteristics in case of <i>TOR1AIP1</i> muscular dystrophy. <i>Clinical Imaging</i> , 2019, 58, 108-113.	0.8	6
43	Essentials of Neonatal-Perinatal Medicine fellowship: an overview. <i>Journal of Perinatology</i> , 2022, 42, 269-276.	0.9	6
44	<i>SMAD4</i> Germline Mutations in Juvenile Polyposis Coli. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 1999, 28, 538-539.	0.9	6
45	Digenic Variants in the <i>FGF21</i> Signaling Pathway Associated with Severe Insulin Resistance and Pseudoacromegaly. <i>Journal of the Endocrine Society</i> , 2020, 4, bvaa138.	0.1	6
46	Biallelic <i>ASCC1</i> variants including a novel intronic variant result in expanded phenotypic spectrum of spinal muscular atrophy with congenital bone fractures 2 ( <i>SMABF2</i> ). <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2190-2197.	0.7	4
47	Distinguishing severe phenotypes associated with pathogenic variants in <i>POLR3A</i> . <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 708-712.	0.7	4
48	Biologic characterization of <i>ABCA3</i> variants in lung tissue from infants and children with <i>ABCA3</i> deficiency. <i>Pediatric Pulmonology</i> , 2022, 57, 1325-1330.	1.0	3
49	ATS Core Curriculum 2016: Part III. Pediatric Pulmonary Medicine. <i>Annals of the American Thoracic Society</i> , 2016, 13, 955-966.	1.5	2
50	A Step toward Treating a Lethal Neonatal Lung Disease. <i>STAT3</i> and Alveolar Capillary Dysplasia. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2019, 200, 961-962.	2.5	2
51	Postmenstrual age at discharge in premature infants with and without ventilatory pattern instability. <i>Journal of Perinatology</i> , 2020, 40, 157-162.	0.9	2
52	Congenital Acute Myeloid Leukemia with Unique Translocation t(11;19)(q23;p13.3). <i>Cureus</i> , 2015, 7, e289.	0.2	2
53	First Steps toward Personalized Therapies for <i>ABCA3</i> Deficiency. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2022, 66, 349-350.	1.4	2
54	Gene variants of the phosphatidylcholine synthesis pathway do not contribute to RDS in the Chinese population. <i>World Journal of Pediatrics</i> , 2018, 14, 52-56.	0.8	1

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55	Basic Genetics and Epigenetics of Childhood Lung Disease. , 2019, , 40-48.e2.		1
56	Whole exome sequencing and functional characterization increase diagnostic yield in siblings with a 46, XY difference of sexual development (DSD). Journal of Steroid Biochemistry and Molecular Biology, 2021, 212, 105908.	1.2	1
57	Respiratory Distress, Flaccid Abdominal Musculature, and Cryptorchidism. NeoReviews, 2011, 12, e55-e58.	0.4	0
58	Blueberry Muffin Rash and Respiratory Distress in a Late Preterm Infant. NeoReviews, 2012, 13, e506-e510.	0.4	0
59	Term Newborn with Abdominal Wall Defect, Ambiguous Genitalia, and Respiratory Distress. NeoReviews, 2013, 14, e211-e215.	0.4	0
60	Case 38: Respiratory Distress, Flaccid Abdominal Musculature, and Cryptorchidism. , 2018, , 267-273.		0
61	Case 28: Blueberry Muffin Rash and Respiratory Distress in a Late-Preterm Infant. , 2018, , 201-208.		0
62	Lethal neonatal respiratory failure due to biallelic variants in BBS1 and monoallelic variant in TTC21B. Pediatric Nephrology, 0, , .	0.9	0