Jennifer A Wambach

List of Publications by Year
in descending order

62	1,816	430442	288905
papers	citations	18 h-index	40 g-index

66	66	66	2632
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Essentials of Neonatal-Perinatal Medicine fellowship: an overview. Journal of Perinatology, 2022, 42, 269-276.	0.9	6
2	Distinguishing severe phenotypes associated with pathogenic variants in POLR3A. American Journal of Medical Genetics, Part A, 2022, 188, 708-712.	0.7	4
3	First Steps toward Personalized Therapies for ABCA3 Deficiency. American Journal of Respiratory Cell and Molecular Biology, 2022, 66, 349-350.	1.4	2
4	A dominant negative variant of RAB5B disrupts maturation of surfactant protein B and surfactant protein C. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	9
5	Biologic characterization of ABCA3 variants in lung tissue from infants and children with ABCA3 deficiency. Pediatric Pulmonology, 2022, 57, 1325-1330.	1.0	3
6	Discovery of a novel <scp>CHD7 CHARGE</scp> syndrome variant by integrated omics analyses. American Journal of Medical Genetics, Part A, 2021, 185, 544-548.	0.7	10
7	Biallelic <scp>ASCC1</scp> variants including a novel intronic variant result in expanded phenotypic spectrum of spinal muscular atrophy with congenital bone fractures 2 (<scp>SMABF2</scp>). American Journal of Medical Genetics, Part A, 2021, 185, 2190-2197.	0.7	4
8	The common ABCA3^{E292V} variant disrupts AT2 cell quality control and increases susceptibility to lung injury and aberrant remodeling. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2021, 321, L291-L307.	1.3	16
9	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
10	Gene Therapy Potential for Genetic Disorders of Surfactant Dysfunction. Frontiers in Genome Editing, 2021, 3, 785829.	2.7	13
11	Postmenstrual age at discharge in premature infants with and without ventilatory pattern instability. Journal of Perinatology, 2020, 40, 157-162.	0.9	2
12	Functional Genomics of ABCA3 Variants. American Journal of Respiratory Cell and Molecular Biology, 2020, 63, 436-443.	1.4	19
13	Childhood rare lung disease in the 21st century: â€œâ€¢omicsâ€•technology advances accelerating discovery. Pediatric Pulmonology, 2020, 55, 1828-1837.	1.0	8
14	Functional characterization of four ATPâ€•binding cassette transporter A3 gene (ABCA3) variants. Human Mutation, 2020, 41, 1298-1307.	1.1	13
15	Phenotypic expansion of KMT2Dâ€related disorder: Beyond Kabuki syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1053-1065.	0.7	23
16	Neuroendocrine Cell Hyperplasia of Infancy. Clinical Score and Comorbidities. Annals of the American Thoracic Society, 2020, 17, 724-728.	1.5	21
17	Digenic Variants in the FGF21 Signaling Pathway Associated with Severe Insulin Resistance and Pseudoacromegaly. Journal of the Endocrine Society, 2020, 4, bvaa138.	0.1	6
18	Basic Genetics and Epigenetics of Childhood Lung Disease. , 2019, , 40-48.e2.		1

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19	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	0.8	6
20	A Step toward Treating a Lethal Neonatal Lung Disease. STAT3 and Alveolar Capillary Dysplasia. American Journal of Respiratory and Critical Care Medicine, 2019, 200, 961-962.	2.5	2
21	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	2.6	59
22	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	1.1	19
23	Phenotype and response to growth hormone therapy in siblings with B4GALT7 deficiency. Bone, 2019, 124, 14-21.	1.4	9
24	CemOrange2 fusions facilitate multifluorophore subcellular imaging in C. elegans. PLoS ONE, 2019, 14, e0214257.	1.1	11
25	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60.	1.8	18
26	Gene variants of the phosphatidylcholine synthesis pathway do not contribute to RDS inÂthe Chinese population. World Journal of Pediatrics, 2018, 14, 52-56.	0.8	1
27	Infants with Atypical Presentations of Alveolar Capillary Dysplasia with Misalignment of the Pulmonary Veins Who Underwent Bilateral Lung Transplantation. Journal of Pediatrics, 2018, 194, 158-164.e1.	0.9	48
28	Neonatal Outcomes Differ after Spontaneous and Indicated Preterm Birth. American Journal of Perinatology, 2018, 35, 494-502.	0.6	8
29	Bi-allelic POLR3A Loss-of-Function Variants Cause Autosomal-Recessive Wiedemann-Rautenstrauch Syndrome. American Journal of Human Genetics, 2018, 103, 968-975.	2.6	43
30	Functional characterization of biallelic RTTN variants identified in an infant with microcephaly, simplified gyral pattern, pontocerebellar hypoplasia, and seizures. Pediatric Research, 2018, 84, 435-441.	1.1	11
31	LINE- andAlu-containing genomic instability hotspot at 16q24.1 associated with recurrent and nonrecurrent CNV deletions causative for ACDMPV. Human Mutation, 2018, 39, 1916-1925.	1.1	14
32	Prevention and Treatment of Respiratory Distress Syndrome in Preterm Neonates. Neonatal Network: NN, 2018, 37, 169-177.	0.1	32
33	Case 38: Respiratory Distress, Flaccid Abdominal Musculature, and Cryptorchidism. , 2018, , 267-273.		0
34	Case 28: Blueberry Muffin Rash and Respiratory Distress in a Late-Preterm Infant. , 2018, , 201-208.		0
35	Outcomes of Lung Transplantation for Infants and Children with Genetic Disorders of Surfactant Metabolism. Journal of Pediatrics, 2017, 184, 157-164.e2.	0.9	66
36	Differentiation of Human Pluripotent Stem Cells into Functional Lung Alveolar Epithelial Cells. Cell Stem Cell, 2017, 21, 472-488.e10.	5.2	406

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37	Survival among children with â€œLethalâ€•congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene (GLDN). Human Mutation, 2017, 38, 1477-1484.	1.1	19
38	Functional Characterization of ATP-Binding Cassette Transporter A3 Mutations from Infants with Respiratory Distress Syndrome. American Journal of Respiratory Cell and Molecular Biology, 2016, 55, 716-721.	1.4	44
39	Accelerating Scientific Advancement for Pediatric Rare Lung Disease Research. Report from a National Institutes of Healthâ€“NHLBI Workshop, September 3 and 4, 2015. Annals of the American Thoracic Society, 2016, 13, 385-393.	1.5	9
40	ATS Core Curriculum 2016: Part III. Pediatric Pulmonary Medicine. Annals of the American Thoracic Society, 2016, 13, 955-966.	1.5	2
41	Genetic Factors Contribute to Risk for Neonatal Respiratory Distress Syndrome among Moderately Preterm, Late Preterm, and Term Infants. Journal of Pediatrics, 2016, 172, 69-74.e2.	0.9	14
42	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. American Journal of Medical Genetics, Part A, 2015, 167, 2966-2974.	0.7	16
43	Remission of Severe Neonatal Diabetes With Very Early Sulfonylurea Treatment. Diabetes Care, 2015, 38, e38-e39.	4.3	15
44	Congenital Acute Myeloid Leukemia with Unique Translocation t(11;19)(q23;p13.3). Cureus, 2015, 7, e289.	0.2	2
45	New clinical practice guidelines on the classification, evaluation and management of childhood interstitial lung disease in infants: what do they mean?. Expert Review of Respiratory Medicine, 2014, 8, 653-655.	1.0	9
46	Two deletions overlapping a distant FOXF1 enhancer unravel the role of lncRNA LINC01081 in etiology of alveolar capillary dysplasia with misalignment of pulmonary veins. American Journal of Medical Genetics, Part A, 2014, 164, 2013-2019.	0.7	46
47	Genotypeâ€“Phenotype Correlations for Infants and Children with ABCA3 Deficiency. American Journal of Respiratory and Critical Care Medicine, 2014, 189, 1538-1543.	2.5	168
48	Synonymous ABCA3 Variants Do Not Increase Risk for Neonatal Respiratory Distress Syndrome. Journal of Pediatrics, 2014, 164, 1316-1321.e3.	0.9	6
49	NovelFOXF1Mutations in Sporadic and Familial Cases of Alveolar Capillary Dysplasia with Misaligned Pulmonary Veins Imply a Role for its DNA Binding Domain. Human Mutation, 2013, 34, 801-811.	1.1	97
50	Term Newborn with Abdominal Wall Defect, Ambiguous Genitalia, and Respiratory Distress. NeoReviews, 2013, 14, e211-e215.	0.4	0
51	An intronic ABCA3 mutation that is responsible for respiratory disease. Pediatric Research, 2012, 71, 633-637.	1.1	46
52	Blueberry Muffin Rash and Respiratory Distress in a Late Preterm Infant. NeoReviews, 2012, 13, e506-e510.	0.4	0
53	Single ABCA3 Mutations Increase Risk for Neonatal Respiratory Distress Syndrome. Pediatrics, 2012, 130, e1575-e1582.	1.0	93
54	Respiratory Distress, Flaccid Abdominal Musculature, and Cryptorchidism. NeoReviews, 2011, 12, e55-e58.	0.4	0

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55	Surfactant Protein-C Promoter Variants Associated With Neonatal Respiratory Distress Syndrome Reduce Transcription. Pediatric Research, 2010, 68, 216-220.	1.1	34
56	Womenâ€™s Lifelong Exposure to Neighborhood Poverty and Low Birth Weight: A Population-Based Study. Maternal and Child Health Journal, 2009, 13, 326-33.	0.7	75
57	Successful sulfonylurea treatment of an insulin-naÃ¯ve neonate with diabetes mellitus due to a KCNJ11 mutation. Pediatric Diabetes, 2009, 11, 286-288.	1.2	35
58	Recombination as a mechanism for sporadic mutation in the surfactant proteinâ€C gene. Pediatric Pulmonology, 2008, 43, 443-450.	1.0	11
59	Population and Disease-Based Prevalence of the Common Mutations Associated With Surfactant Deficiency. Pediatric Research, 2008, 63, 645-649.	1.1	94
60	Gastrointestinal polyps in children: Advances in molecular genetics, diagnosis, and management. Journal of Pediatrics, 2001, 138, 621-628.	0.9	55
61	SMAD4 Germline Mutations in Juvenile Polyposis Coli. Journal of Pediatric Gastroenterology and Nutrition, 1999, 28, 538-539.	0.9	6
62	Lethal neonatal respiratory failure due to biallelic variants in BBS1 and monoallelic variant in TTC21B. Pediatric Nephrology, 0, , .	0.9	0