

Debra Ellyn Weese-Mayer

List of Publications by Year in descending order

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Version: 2024-02-01

83
papers

5,098
citations

94433

37
h-index

91884

69
g-index

93
all docs

93
docs citations

93
times ranked

3406
citing authors

#	ARTICLE	IF	CITATIONS
1	Evolution of physiologic and autonomic phenotype in rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation over a decade from age at diagnosis. <i>Journal of Clinical Sleep Medicine</i> , 2022, 18, 937-944.	2.6	4
2	Cerebral Autoregulation during Orthostatic Challenge in Congenital Central Hypoventilation Syndrome. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2022, 205, 340-349.	5.6	6
3	Neurocognitive monitoring in congenital central hypoventilation syndrome with the <i>NIH Toolbox</i>. <i>Pediatric Pulmonology</i> , 2022, 57, 2040-2047.	2.0	8
4	Kangaroo father care: A pilot feasibility study of physiologic, biologic, and psychosocial measures to capture the effects of fatherâ€™ infant and motherâ€™ infant skinâ€™toâ€™skin contact in the Neonatal Intensive Care Unit. <i>Developmental Psychobiology</i> , 2021, 63, 1521-1533.	1.6	15
5	The privilege and responsibility of caring for patients with rare genetic disorders. <i>Clinical Autonomic Research</i> , 2021, 31, 55-56.	2.5	0
6	Development of a Heart Rate Variability Risk Score to Predict Organ Dysfunction and Death in Critically Ill Children. <i>Pediatric Critical Care Medicine</i> , 2021, 22, e437-e447.	0.5	16
7	Paired-like homeobox gene (PHOX2B) nonpolyalanine repeat expansion mutations (NPARMs): genotypeâ€™ phenotype correlation in congenital central hypoventilation syndrome (CCHS). <i>Genetics in Medicine</i> , 2021, 23, 1656-1663.	2.4	16
8	Cerebral cortical-autonomic connectivity in newborns: a first step to determine the autonomic signatures with advancing age?. <i>Clinical Autonomic Research</i> , 2021, 31, 359-360.	2.5	0
9	Wireless, Skinâ€™Interfaced Devices for Pediatric Critical Care: Application to Continuous, Noninvasive Blood Pressure Monitoring. <i>Advanced Healthcare Materials</i> , 2021, 10, e2100383.	7.6	33
10	Autonomic Nervous System Dysfunction Is Associated With Re-hospitalization in Pediatric Septic Shock Survivors. <i>Frontiers in Pediatrics</i> , 2021, 9, 745844.	1.9	2
11	Pupillometry measures of autonomic nervous system regulation with advancing age in a healthy pediatric cohort. <i>Clinical Autonomic Research</i> , 2020, 30, 43-51.	2.5	22
12	Machine learning mortality classification in clinical documentation with increased accuracy in visualâ€™based analyses. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2020, 109, 1346-1353.	1.5	5
13	The Pathophysiology of Rett Syndrome With a Focus on Breathing Dysfunctions. <i>Physiology</i> , 2020, 35, 375-390.	3.1	20
14	A wireless, skin-interfaced biosensor for cerebral hemodynamic monitoring in pediatric care. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 31674-31684.	7.1	55
15	Diurnal variation in autonomic regulation among patients with genotyped Rett syndrome. <i>Journal of Medical Genetics</i> , 2020, 57, 786-793.	3.2	17
16	Skin-interfaced biosensors for advanced wireless physiological monitoring in neonatal and pediatric intensive-care units. <i>Nature Medicine</i> , 2020, 26, 418-429.	30.7	272
17	Airway Obstruction during Sleep due to Diaphragm Pacing Precludes Decannulation in Young Children with CCHS. <i>Respiration</i> , 2019, 98, 263-267.	2.6	15
18	Congenital central hypoventilation syndrome: Severe disease caused by coâ€™occurrence of two PHOX2B variants inherited separately from asymptomatic family members. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 503-506.	1.2	10

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19	Binodal, wireless epidermal electronic systems with in-sensor analytics for neonatal intensive care. <i>Science</i> , 2019, 363, .	12.6	521
20	Pre-Vent: the prematurity-related ventilatory control study. <i>Pediatric Research</i> , 2019, 85, 769-776.	2.3	33
21	Congenital central hypoventilation syndrome: Broader cognitive deficits revealed by parent controls. <i>Pediatric Pulmonology</i> , 2018, 53, 492-497.	2.0	16
22	Perioperative anesthetic management of children with congenital central hypoventilation syndrome and rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation undergoing thoracoscopic phrenic nerve-diaphragm pacemaker implantation. <i>Paediatric Anaesthesia</i> , 2018, 28, 963-973.	1.1	8
23	Autonomic Nervous System Dysfunction in Pediatric Sepsis. <i>Frontiers in Pediatrics</i> , 2018, 6, 280.	1.9	43
24	ROHHAD and Prader-Willi syndrome (PWS): clinical and genetic comparison. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 124.	2.7	27
25	Chronic nausea and orthostatic intolerance: Diagnostic utility of orthostatic challenge duration, Nausea Profile Questionnaire, and neurohumoral measures. <i>Neurogastroenterology and Motility</i> , 2018, 30, e13433.	3.0	6
26	Mutations in <i>MYO1H</i> cause a recessive form of central hypoventilation with autonomic dysfunction. <i>Journal of Medical Genetics</i> , 2017, 54, 754-761.	3.2	21
27	Congenital central hypoventilation syndrome: a bedside-to-bench success story for advancing early diagnosis and treatment and improved survival and quality of life. <i>Pediatric Research</i> , 2017, 81, 192-201.	2.3	51
28	Determining peripheral skin temperature: subjective versus objective measurements. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2016, 105, e126-e131.	1.5	17
29	Stillbirth: Correlations between Brain Injury and Placental Pathology. <i>Pediatric and Developmental Pathology</i> , 2016, 19, 237-243.	1.0	18
30	Congenital Central Hypoventilation Syndrome. <i>Chest</i> , 2016, 149, 809-815.	0.8	44
31	Congenital central hypoventilation syndrome (CCHS): Circadian temperature variation. <i>Pediatric Pulmonology</i> , 2016, 51, 300-307.	2.0	16
32	Absence of mutations in <i>HCRT</i> , <i>HCRTR1</i> and <i>HCRTR2</i> in patients with ROHHAD. <i>Respiratory Physiology and Neurobiology</i> , 2016, 221, 59-63.	1.6	19
33	Stillbirth evaluation: a stepwise assessment of placental pathology and autopsy. <i>American Journal of Obstetrics and Gynecology</i> , 2016, 214, 115.e1-115.e6.	1.3	40
34	Rapid-Onset Obesity with Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation (ROHHAD): exome sequencing of trios, monozygotic twins and tumours. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 103.	2.7	45
35	Treatment of neuroblastoma in congenital central hypoventilation syndrome with a <i>PHOX2B</i> polyalanine repeat expansion mutation: New twist on a neurocristopathy syndrome. <i>Pediatric Blood and Cancer</i> , 2015, 62, 2007-2010.	1.5	12
36	Rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation (ROHHAD): Response to ventilatory challenges. <i>Pediatric Pulmonology</i> , 2015, 50, 1336-1345.	2.0	30

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37	Residual chemosensitivity to ventilatory challenges in genotyped congenital central hypoventilation syndrome. <i>Journal of Applied Physiology</i> , 2014, 116, 439-450.	2.5	39
38	Congenital Central Hypoventilation Syndrome. <i>Clinics in Chest Medicine</i> , 2014, 35, 535-545.	2.1	26
39	Hypoventilation Syndromes of Infancy, Childhood, and Adulthood. <i>Sleep Medicine Clinics</i> , 2014, 9, 425-439.	2.6	2
40	Congenital Central Hypoventilation Syndrome and Sudden Infant Death Syndrome: Disorders of Autonomic Regulation. <i>Seminars in Pediatric Neurology</i> , 2013, 20, 44-55.	2.0	25
41	Commentary: Rapid-onset Obesity with Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation (ROHHAD): Remember Your ABCs (Airway, Breathing, Circulation). <i>Journal of the Canadian Academy of Child and Adolescent Psychiatry</i> , 2013, 22, 238-9.	0.6	4
42	Pupillometry in congenital central hypoventilation syndrome (CCHS): quantitative evidence of autonomic nervous system dysregulation. <i>Pediatric Research</i> , 2012, 71, 280-285.	2.3	41
43	Variable human phenotype associated with novel deletions of the <i>PHOX2B</i> gene. <i>Pediatric Pulmonology</i> , 2012, 47, 153-161.	2.0	54
44	Germline mosaicism of <i>PHOX2B</i> mutation accounts for familial recurrence of congenital central hypoventilation syndrome (CCHS). <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2297-2301.	1.2	23
45	Rapid-Onset Obesity With Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation: Analysis of Hypothalamic and Autonomic Candidate Genes. <i>Pediatric Research</i> , 2011, 70, 375-378.	2.3	66
46	Monozygotic Twins Discordant for ROHHAD Phenotype. <i>Pediatrics</i> , 2011, 128, e711-e715.	2.1	47
47	Carbon dioxide chemoreception and hypoventilation syndromes with autonomic dysregulation. <i>Journal of Applied Physiology</i> , 2010, 108, 979-988.	2.5	35
48	Congenital central hypoventilation syndrome: Neurocognitive functioning in school age children. <i>Pediatric Pulmonology</i> , 2010, 45, 92-98.	2.0	56
49	Comparison of PHOX2B Testing Methods in the Diagnosis of Congenital Central Hypoventilation Syndrome and Mosaic Carriers. <i>Diagnostic Molecular Pathology</i> , 2010, 19, 224-231.	2.1	23
50	An Official ATS Clinical Policy Statement: Congenital Central Hypoventilation Syndrome. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2010, 181, 626-644.	5.6	433
51	Later-onset congenital central hypoventilation syndrome due to a heterozygous 24â€polyalanine repeat expansion mutation in the <i>PHOX2B</i> gene. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2009, 98, 192-195.	1.5	45
52	Congenital central hypoventilation syndrome from past to future: Model for translational and transitional autonomic medicine. <i>Pediatric Pulmonology</i> , 2009, 44, 521-535.	2.0	99
53	Congenital central hypoventilation syndrome: <i>PHOX2B</i> genotype determines risk for sudden death. <i>Pediatric Pulmonology</i> , 2008, 43, 77-86.	2.0	105
54	Sudden infant death syndrome: the genetic segue?. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2008, 97, 846-847.	1.5	4

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55	Congenital central hypoventilation syndrome (CCHS) and sudden infant death syndrome (SIDS): Kindred disorders of autonomic regulation. <i>Respiratory Physiology and Neurobiology</i> , 2008, 164, 38-48.	1.6	51
56	Rapid-Onset Obesity With Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation Presenting in Childhood. <i>Pediatrics</i> , 2007, 120, e179-e188.	2.1	175
57	Vagal and sympathetic heart rate and blood pressure control in adult onset PHOX2B mutationâ€“confirmed congenital central hypoventilation syndrome. <i>Clinical Autonomic Research</i> , 2007, 17, 177-185.	2.5	28
58	PHOX2B Mutationâ€“confirmed Congenital Central Hypoventilation Syndrome. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2006, 174, 923-927.	5.6	125
59	Congenital Central Hypoventilation Syndrome. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2006, 174, 1139-1144.	5.6	238
60	Adult Identified with Congenital Central Hypoventilation Syndromeâ€“Mutation in PHOX2b Gene and Late-Onset CHS. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2005, 171, 88-88.	5.6	80
61	Sudden infant death syndrome: Association with a promoter polymorphism of the serotonin transporter gene. <i>American Journal of Medical Genetics Part A</i> , 2003, 117A, 268-274.	2.4	118
62	Association of the serotonin transporter gene with sudden infant death syndrome: A haplotype analysis. <i>American Journal of Medical Genetics Part A</i> , 2003, 122A, 238-245.	2.4	94
63	Idiopathic congenital central hypoventilation syndrome: Analysis of genes pertinent to early autonomic nervous system embryologic development and identification of mutations in PHOX2b. <i>American Journal of Medical Genetics Part A</i> , 2003, 123A, 267-278.	2.4	335
64	Idiopathic congenital central hypoventilation syndrome: Evaluation of brain-derived neurotrophic factor genomic DNA sequence variation. <i>American Journal of Medical Genetics Part A</i> , 2002, 107, 306-310.	2.4	63
65	Idiopathic congenital central hypoventilation syndrome: the next generation. <i>American Journal of Medical Genetics Part A</i> , 2002, 112, 46-50.	2.4	52
66	Case/control family study of autonomic nervous system dysfunction in idiopathic congenital central hypoventilation syndrome. <i>American Journal of Medical Genetics Part A</i> , 2001, 100, 237-245.	2.4	83
67	Genetic segregation analysis of autonomic nervous system dysfunction in families of probands with idiopathic congenital central hypoventilation syndrome. <i>American Journal of Medical Genetics Part A</i> , 2001, 100, 229-236.	2.4	49
68	Cardiac rhythm disturbances among children with idiopathic congenital central hypoventilation syndrome. , 2000, 29, 351-358.		58
69	Uterine Position Determines the Extent of Dopamine Reduction after Chronic Prenatal Cocaine Exposure. <i>Annals of the New York Academy of Sciences</i> , 1998, 844, 314-323.	3.8	6
70	Congenital central hypoventilation syndrome: Mutation analysis of the receptor tyrosine kinase RET. , 1996, 63, 603-609.		45
71	Endothelinâ€“3 frameshift mutation in congenital central hypoventilation syndrome. <i>Nature Genetics</i> , 1996, 13, 395-396.	21.4	89
72	Congenital central hypoventilation syndrome: Cardiorespiratory responses to moderate exercise, simulating daily activity. <i>Pediatric Pulmonology</i> , 1995, 20, 89-93.	2.0	52

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73	Congenital central hypoventilation syndrome: Inheritance and relation to sudden infant death syndrome. <i>American Journal of Medical Genetics Part A</i> , 1993, 47, 360-367.	2.4	78
74	Polysomnography in obese children with a history of sleep-associated breathing disorders. <i>Pediatric Pulmonology</i> , 1993, 16, 124-129.	2.0	182
75	Drug Screen Technique Determines Prevalence of Cocaine Use Among Pregnant Women. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 1992, 1, 148-152.	1.5	0
76	Diaphragm pacing in infants and children. <i>Journal of Pediatrics</i> , 1992, 120, 1-8.	1.8	83
77	Congenital central hypoventilation syndrome: Diagnosis, management, and long-term outcome in thirty-two children. <i>Journal of Pediatrics</i> , 1992, 120, 381-387.	1.8	195
78	Neuropsychologic abnormalities in children with congenital central hypoventilation syndrome. <i>Journal of Pediatrics</i> , 1992, 120, 388-393.	1.8	52
79	Effect of prenatal cocaine on respiration, heart rate, and sudden infant death syndrome. <i>Pediatric Pulmonology</i> , 1991, 11, 328-334.	2.0	60
80	Breathing Control Disorders in Infants and Children. <i>Hospital Practice (1995)</i> , 1990, 25, 82-103.	1.0	7
81	Comparison of transthoracic impedance/heart rate monitoring and pulse oximetry for patients using diaphragm pacemakers. <i>Pediatric Pulmonology</i> , 1990, 8, 29-32.	2.0	10
82	Magnetic Resonance Imaging and Computerized Tomography in Central Hypoventilation. <i>The American Review of Respiratory Disease</i> , 1988, 137, 393-398.	2.9	64
83	Environmental Temperature Extremes: Feasibility Study of Effect on Pediatric Health. , 0, , .		0