

Marshall Summar

List of Publications by Year in descending order

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Version: 2024-02-01

104
papers

5,167
citations

71102

41
h-index

95266

68
g-index

106
all docs

106
docs citations

106
times ranked

6249
citing authors

#	ARTICLE	IF	CITATIONS
1	The Rare Disease Research Scholars Program: A training curriculum for clinical researchers with mixed methods evaluation study. <i>Translational Science of Rare Diseases</i> , 2022, 6, 1-11.	1.5	1
2	A machine learning-based screening tool for genetic syndromes in children – Authors' reply. <i>The Lancet Digital Health</i> , 2022, 4, e296.	12.3	0
3	Acylcarnitines and Genetic Variation in Fat Oxidation Genes in HIV-infected, Antiretroviral-treated Children With and Without Myopathy. <i>Pediatric Infectious Disease Journal</i> , 2022, 41, e306-e311.	2.0	0
4	Rapid deployment of a telemedicine care model for genetics and metabolism during COVID-19. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 68-72.	1.2	36
5	Development and evaluation of a machine learning-based point-of-care screening tool for genetic syndromes in children: a multinational retrospective study. <i>The Lancet Digital Health</i> , 2021, 3, e635-e643.	12.3	38
6	Folic acid, either solely or combined with L-citrulline, improves NO signaling and ameliorates chronic hypoxia-induced pulmonary hypertension in newborn pigs. <i>Physiological Reports</i> , 2021, 9, e15096.	1.7	1
7	Combined L-citrulline and tetrahydrobiopterin therapy improves NO signaling and ameliorates chronic hypoxia-induced pulmonary hypertension in newborn pigs. <i>American Journal of Physiology - Lung Cellular and Molecular Physiology</i> , 2020, 318, L762-L772.	2.9	8
8	Response to letter, broken bones, and irresponsible testimony: Enough is enough already: The flawed Ehlers-Danlos syndrome infant fragility theory should not rule. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2335-2337.	1.2	3
9	Invited commentary: His life was lost but his heart still beats: In honor of children harmed by child abuse. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2329-2332.	1.2	3
10	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. <i>Annals of Neurology</i> , 2019, 86, 116-128.	5.3	42
11	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders – A successful strategy for clinical research of rare diseases. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 93-106.	3.6	35
12	Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 150-158.	1.2	40
13	A phase 1 dose-finding study of intravenous L-citrulline in sickle cell disease: a potential novel therapy for sickle cell pain crisis. <i>British Journal of Haematology</i> , 2019, 184, 634-636.	2.5	10
14	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders – a successful strategy for clinical research of rare diseases. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 93.	3.6	4
15	Williams-Beuren syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1128-1136.	1.2	55
16	Cover Image, Volume 176A, Number 5, May 2018. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, .	1.2	0
17	22q11.2 deletion syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 879-888.	1.2	103
18	Down syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 42-53.	1.2	75

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19	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
20	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	1.2	68
21	Identification of dysmorphic syndromes using landmark-specific local texture descriptors. , 2016, , .		26
22	Tetrahydrobiopterin oral therapy recouples eNOS and ameliorates chronic hypoxia-induced pulmonary hypertension in newborn pigs. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2016, 311, L743-L753.	2.9	26
23	Climbing the Ladder: Experience with Developing a Large Group Genetic Counselor Career Ladder at Children's National Health System. Journal of Genetic Counseling, 2016, 25, 644-648.	1.6	7
24	Preoperative evaluation and comprehensive risk assessment for children with Down syndrome. Paediatric Anaesthesia, 2016, 26, 356-362.	1.1	61
25	Acylcarnitine Profiles in HIV-Exposed, Uninfected Neonates in the United States. AIDS Research and Human Retroviruses, 2016, 32, 339-348.	1.1	14
26	Targeted Echocardiographic Screening for Latent Rheumatic Heart Disease in Northern Uganda: Evaluating Familial Risk Following Identification of an Index Case. PLoS Neglected Tropical Diseases, 2016, 10, e0004727.	3.0	22
27	Short-term follow-up systems for positive newborn screens in the Washington Metropolitan Area and the United States. Molecular Genetics and Metabolism, 2015, 116, 226-230.	1.1	3
28	Constrained local model with independent component analysis and kernel density estimation: Application to down syndrome detection. , 2015, , .		3
29	Simple and inexpensive quantification of ammonia in whole blood. Molecular Genetics and Metabolism, 2015, 115, 95-100.	1.1	17
30	Rescue Treatment with L-Citrulline Inhibits Hypoxia-Induced Pulmonary Hypertension in Newborn Pigs. American Journal of Respiratory Cell and Molecular Biology, 2015, 53, 255-264.	2.9	50
31	Malignancy in Noonan syndrome and related disorders. Clinical Genetics, 2015, 88, 516-522.	2.0	38
32	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. Journal of Inherited Metabolic Disease, 2015, 38, 1059-1074.	3.6	175
33	A novel mutation in the promoter of RARS2 causes pontocerebellar hypoplasia in two siblings. Journal of Human Genetics, 2015, 60, 363-369.	2.3	26
34	Blood ammonia and glutamine as predictors of hyperammonemic crises in patients with urea cycle disorder. Genetics in Medicine, 2015, 17, 561-568.	2.4	30
35	Sodium-Coupled Neutral Amino Acid Transporter 1 (SNAT1) Modulates L-Citrulline Transport and Nitric Oxide (NO) Signaling in Piglet Pulmonary Arterial Endothelial Cells. PLoS ONE, 2014, 9, e85730.	2.5	16
36	Ensemble learning for the detection of facial dysmorphology. , 2014, 2014, 754-7.		8

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37	Genetic susceptibility to endomyocardial fibrosis. <i>Global Cardiology Science & Practice</i> , 2014, 2014, 60.	0.4	12
38	Liver transplantation for pediatric metabolic disease. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 418-427.	1.1	105
39	L-citrulline provides a novel strategy for treating chronic pulmonary hypertension in newborn infants. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2014, 103, 1019-1026.	1.5	36
40	Mind the Gap. <i>Molecular Genetics and Metabolism</i> , 2014, 113, 1.	1.1	3
41	A longitudinal study of urea cycle disorders. <i>Molecular Genetics and Metabolism</i> , 2014, 113, 127-130.	1.1	153
42	Save the biochemical geneticists!. <i>Molecular Genetics and Metabolism</i> , 2014, 113, 2-3.	1.1	4
43	Digital facial dysmorphology for genetic screening: Hierarchical constrained local model using ICA. <i>Medical Image Analysis</i> , 2014, 18, 699-710.	11.6	70
44	Molecular Characterization of Carbamoyl-Phosphate Synthetase (CPS1) Deficiency Using Human Recombinant CPS1 as a Key Tool. <i>Human Mutation</i> , 2013, 34, 1149-1159.	2.5	34
45	The incidence of urea cycle disorders. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 179-180.	1.1	232
46	Down syndrome detection from facial photographs using machine learning techniques. <i>Proceedings of SPIE</i> , 2013, , .	0.8	13
47	Automated down syndrome detection using facial photographs. , 2013, 2013, 3670-3.		8
48	Abnormal Newborn Screens and Acylcarnitines in HIV-exposed and ARV-exposed Infants. <i>Pediatric Infectious Disease Journal</i> , 2013, 32, 146-150.	2.0	19
49	Hierarchical Constrained Local Model Using ICA and Its Application to Down Syndrome Detection. <i>Lecture Notes in Computer Science</i> , 2013, 16, 222-229.	1.3	19
50	Significant Differences in Markers of Oxidant Injury between Idiopathic and Bronchopulmonary-Dysplasia-Associated Pulmonary Hypertension in Children. <i>Pulmonary Medicine</i> , 2012, 2012, 1-6.	1.9	6
51	Prolonged hypoxia augments l-citrulline transport by System A in the newborn piglet pulmonary circulation. <i>Cardiovascular Research</i> , 2012, 95, 375-384.	3.8	13
52	Urinary phenylacetylglutamine as dosing biomarker for patients with urea cycle disorders. <i>Molecular Genetics and Metabolism</i> , 2012, 107, 308-314.	1.1	38
53	Chronic management and health supervision of individuals with propionic acidemia. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 26-33.	1.1	75
54	Natural history of propionic acidemia. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 5-9.	1.1	120

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55	Acute management of propionic acidemia. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 16-25.	1.1	96
56	Neurologic considerations in propionic acidemia. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 10-15.	1.1	76
57	Peptide tyrosine tyrosine levels are increased in patients with urea cycle disorders. <i>Molecular Genetics and Metabolism</i> , 2012, 106, 39-42.	1.1	4
58	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. <i>Human Mutation</i> , 2012, 33, 165-179.	2.5	45
59	Shift Work in Nurses: Contribution of Phenotypes and Genotypes to Adaptation. <i>PLoS ONE</i> , 2011, 6, e18395.	2.5	137
60	Molecular defects in human carbamoyl phosphate synthetase I: mutational spectrum, diagnostic and protein structure considerations. <i>Human Mutation</i> , 2011, 32, 579-589.	2.5	67
61	Vaccines Are Not Associated With Metabolic Events in Children With Urea Cycle Disorders. <i>Pediatrics</i> , 2011, 127, e1147-e1153.	2.1	31
62	Evaluation of Immunization Rates and Safety Among Children With Inborn Errors of Metabolism. <i>Pediatrics</i> , 2011, 127, e1139-e1146.	2.1	38
63	Requirement of argininosuccinate lyase for systemic nitric oxide production. <i>Nature Medicine</i> , 2011, 17, 1619-1626.	30.7	189
64	Understanding the role of NOS-3 in ventilator-induced lung injury: don't take NO for an answer. <i>American Journal of Physiology - Lung Cellular and Molecular Physiology</i> , 2010, 299, L147-L149.	2.9	4
65	L-Citrulline Attenuates Arrested Alveolar Growth and Pulmonary Hypertension in Oxygen-Induced Lung Injury in Newborn Rats. <i>Pediatric Research</i> , 2010, 68, 519-525.	2.3	74
66	Establishing a consortium for the study of rare diseases: The Urea Cycle Disorders Consortium. <i>Molecular Genetics and Metabolism</i> , 2010, 100, S97-S105.	1.1	73
67	Effects of Meals High in Carbohydrate, Protein, and Fat on Ghrelin and Peptide YY Secretion in Prepubertal Children. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 4463-4471.	3.6	55
68	Genetic variation in the urea cycle: a model resource for investigating key candidate genes for common diseases. <i>Human Mutation</i> , 2009, 30, 56-60.	2.5	43
69	Genetic variants of GSNOR and ADRB2 influence response to albuterol in African-American children with severe asthma. <i>Pediatric Pulmonology</i> , 2009, 44, 649-654.	2.0	61
70	Quantitative RT-PCR comparison of the urea and nitric oxide cycle gene transcripts in adult human tissues. <i>Molecular Genetics and Metabolism</i> , 2009, 97, 121-127.	1.1	32
71	Comparison of alterations in amino acids content in cultured astrocytes or neurons exposed to methylmercury separately or in co-culture. <i>Neurochemistry International</i> , 2009, 55, 136-142.	3.8	17
72	Genetic Variation in Complement Component 2 of the Classical Complement Pathway is Associated With Increased Mortality and Infection: A Study of 627 Patients With Trauma. <i>Journal of Trauma</i> , 2009, 66, 1265-1272.	2.3	11

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73	Recommendations for locus-specific databases and their curation. <i>Human Mutation</i> , 2008, 29, 2-5.	2.5	59
74	Cross-sectional multicenter study of patients with urea cycle disorders in the United States. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 397-402.	1.1	189
75	Genetic Differences in Human Circadian Clock Genes among Worldwide Populations. <i>Journal of Biological Rhythms</i> , 2008, 23, 330-340.	2.6	108
76	Population screening in a Druze community: the challenge and the reward. <i>Genetics in Medicine</i> , 2008, 10, 903-909.	2.4	37
77	L-citrulline Inhibits Chronic Hypoxia-Induced Pulmonary Hypertension in Newborn Piglets. <i>FASEB Journal</i> , 2008, 22, 1209.12.	0.5	0
78	Genetic variation in the mitochondrial enzyme carbamyl-phosphate synthetase I predisposes children to increased pulmonary artery pressure following surgical repair of congenital heart defects: A validated genetic association study. <i>Mitochondrion</i> , 2007, 7, 204-210.	3.4	33
79	Pharmacokinetics and safety of intravenously administered citrulline in children undergoing congenital heart surgery: Potential therapy for postoperative pulmonary hypertension. <i>Journal of Thoracic and Cardiovascular Surgery</i> , 2007, 134, 319-326.	0.8	61
80	Nitric oxide precursors and congenital heart surgery: A randomized controlled trial of oral citrulline. <i>Journal of Thoracic and Cardiovascular Surgery</i> , 2006, 132, 58-65.	0.8	83
81	Mitochondrial haplogroups and peripheral neuropathy during antiretroviral therapy: an adult AIDS clinical trials group study. <i>Aids</i> , 2005, 19, 1341-1349.	2.2	129
82	The hemochromatosis C282Y allele: a risk factor for hepatic veno-occlusive disease after hematopoietic stem cell transplantation. <i>Bone Marrow Transplantation</i> , 2005, 35, 1155-1164.	2.4	37
83	Metabolic basis of pediatric heart disease. <i>Progress in Pediatric Cardiology</i> , 2005, 20, 143-159.	0.4	6
84	Late onset N-acetylglutamate synthase deficiency caused by hypomorphic alleles. <i>Human Mutation</i> , 2005, 25, 293-298.	2.5	37
85	Nutritional Management of Urea Cycle Disorders. <i>Critical Care Clinics</i> , 2005, 21, S27-S35.	2.6	42
86	Unmasked Adult-Onset Urea Cycle Disorders in the Critical Care Setting. <i>Critical Care Clinics</i> , 2005, 21, S1-S8.	2.6	120
87	Connecting the dots between genes, biochemistry, and disease susceptibility: systems biology modeling in human genetics. <i>Molecular Genetics and Metabolism</i> , 2005, 84, 104-111.	1.1	30
88	Urea Cycle Disorders: Clinical Presentation Outside the Newborn Period. <i>Critical Care Clinics</i> , 2005, 21, S9-S17.	2.6	78
89	Relationship Between Carbamoyl-Phosphate Synthetase Genotype and Systemic Vascular Function. <i>Hypertension</i> , 2004, 43, 186-191.	2.7	56
90	Multilocus Analysis of Hypertension: A Hierarchical Approach. <i>Human Heredity</i> , 2004, 57, 28-38.	0.8	146

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91	Characterization of genomic structure and polymorphisms in the human carbamyl phosphate synthetase I gene. <i>Gene</i> , 2003, 311, 51-57.	2.2	48
92	Effect of cardiopulmonary bypass on urea cycle intermediates and nitric oxide levels after congenital heart surgery. <i>Journal of Pediatrics</i> , 2003, 142, 26-30.	1.8	59
93	Redox-sensitive interaction between KIAA0132 and Nrf2 mediates indomethacin-induced expression of β -glutamylcysteine synthetase. <i>Free Radical Biology and Medicine</i> , 2002, 32, 650-662.	2.9	72
94	Uniparental disomy of chromosome 2 resulting in lethal trifunctional protein deficiency due to homozygous β -subunit mutations. <i>Human Mutation</i> , 2002, 20, 447-451.	2.5	36
95	Current strategies for the management of neonatal urea cycle disorders. <i>Journal of Pediatrics</i> , 2001, 138, S30-S39.	1.8	139
96	Neonatal Pulmonary Hypertension. <i>New England Journal of Medicine</i> , 2001, 344, 1832-1838.	27.0	202
97	Diagnostic dilemma caused by overlapping features of Prader-Willi syndrome and trisomy 18 during infancy. <i>Journal of Pediatrics</i> , 2000, 136, 135-136.	1.8	4
98	Effective hemodialysis and hemofiltration driven by an extracorporeal membrane oxygenation pump in infants with hyperammonemia. <i>Journal of Pediatrics</i> , 1996, 128, 379-382.	1.8	57
99	Recurrent mutations in the vasopressin-neurophysin II gene cause autosomal dominant neurohypophyseal diabetes insipidus. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1996, 81, 2328-2334.	3.6	14
100	Assignment of the gene (GLCLC) that encodes the heavy subunit of γ -glutamylcysteine synthetase to human chromosome 6. <i>Cytogenetic and Genome Research</i> , 1995, 70, 278-279.	1.1	36
101	Molecular Cloning, Sequencing, Chromosomal Localization, and Tissue Distribution of the Human Na ⁺ /H-Exchanger (SLC9A2). <i>Genomics</i> , 1995, 30, 25-30.	2.9	23
102	Prospective versus clinical diagnosis and therapy of acute neonatal hyperammonaemia in two sisters with carbamyl phosphate synthetase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 1992, 15, 269-277.	3.6	26
103	Linkage analysis of the human dopamine β -hydroxylase gene. <i>Genomics</i> , 1991, 10, 493-495.	2.9	21
104	Linkage Relationships of Human Arginine Vasopressin-Neurophysin-II and Oxytocin-Neurophysin-I to Prodynorphin and Other Loci on Chromosome 20. <i>Molecular Endocrinology</i> , 1990, 4, 947-950.	3.7	36