Marshall Summar

List of Publications by Year
in descending order

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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 incidence of urea cycle disorders. Molecular Genetics and Metabolism, 2013, 110, 179-180.	1.1	232
2	Neonatal Pulmonary Hypertension. New England Journal of Medicine, 2001, 344, 1832-1838.	27.0	202
3	Cross-sectional multicenter study of patients with urea cycle disorders in the United States. Molecular Genetics and Metabolism, 2008, 94, 397-402.	1.1	189
4	Requirement of argininosuccinate lyase for systemic nitric oxide production. Nature Medicine, 2011, 17, 1619-1626.	30.7	189
5	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
6	A longitudinal study of urea cycle disorders. Molecular Genetics and Metabolism, 2014, 113, 127-130.	1.1	153
7	Multilocus Analysis of Hypertension: A Hierarchical Approach. Human Heredity, 2004, 57, 28-38.	0.8	146
8	Current strategies for the management of neonatal urea cycle disorders. Journal of Pediatrics, 2001, 138, S30-S39.	1.8	139
9	Shift Work in Nurses: Contribution of Phenotypes and Genotypes to Adaptation. PLoS ONE, 2011, 6, e18395.	2.5	137
10	Mitochondrial haplogroups and peripheral neuropathy during antiretroviral therapy: an adult AIDS clinical trials group study. Aids, 2005, 19, 1341-1349.	2.2	129
11	Unmasked Adult-Onset Urea Cycle Disorders in the Critical Care Setting. Critical Care Clinics, 2005, 21, S1-S8.	2.6	120
12	Natural history of propionic acidemia. Molecular Genetics and Metabolism, 2012, 105, 5-9.	1.1	120
13	Genetic Differences in Human Circadian Clock Genes among Worldwide Populations. Journal of Biological Rhythms, 2008, 23, 330-340.	2.6	108
14	Liver transplantation for pediatric metabolic disease. Molecular Genetics and Metabolism, 2014, 111, 418-427.	1.1	105
15	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	1.2	103
16	Acute management of propionic acidemia. Molecular Genetics and Metabolism, 2012, 105, 16-25.	1.1	96
17	Nitric oxide precursors and congenital heart surgery: A randomized controlled trial of oral citrulline. Journal of Thoracic and Cardiovascular Surgery, 2006, 132, 58-65.	0.8	83
18	Urea Cycle Disorders: Clinical Presentation Outside the Newborn Period. Critical Care Clinics, 2005, 21, S9-S17.	2.6	78

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19	Neurologic considerations in propionic acidemia. Molecular Genetics and Metabolism, 2012, 105, 10-15.	1.1	76
20	Chronic management and health supervision of individuals with propionic acidemia. Molecular Genetics and Metabolism, 2012, 105, 26-33.	1.1	75
21	Down syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 42-53.	1.2	75
22	L-Citrulline Attenuates Arrested Alveolar Growth and Pulmonary Hypertension in Oxygen-Induced Lung Injury in Newborn Rats. Pediatric Research, 2010, 68, 519-525.	2.3	74
23	Establishing a consortium for the study of rare diseases: The Urea Cycle Disorders Consortium. Molecular Genetics and Metabolism, 2010, 100, S97-S105.	1.1	73
24	Redox-sensitive interaction between KIAA0132 and Nrf2 mediates indomethacin-induced expression of Î³-glutamylcysteine synthetase. Free Radical Biology and Medicine, 2002, 32, 650-662.	2.9	72
25	Digital facial dysmorphology for genetic screening: Hierarchical constrained local model using ICA. Medical Image Analysis, 2014, 18, 699-710.	11.6	70
26	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	1.2	68
27	Molecular defects in human carbamoy phosphate synthetase I: mutational spectrum, diagnostic and protein structure considerations. Human Mutation, 2011, 32, 579-589.	2.5	67
28	Pharmacokinetics and safety of intravenously administered citrulline in children undergoing congenital heart surgery: Potential therapy for postoperative pulmonary hypertension. Journal of Thoracic and Cardiovascular Surgery, 2007, 134, 319-326.	0.8	61
29	Genetic variants of GSNOR and ADRB2 influence response to albuterol in Africanâ€American children with severe asthma. Pediatric Pulmonology, 2009, 44, 649-654.	2.0	61
30	Preoperative evaluation and comprehensive risk assessment for children with <scp>D</scp>own syndrome. Paediatric Anaesthesia, 2016, 26, 356-362.	1.1	61
31	Effect of cardiopulmonary bypass on urea cycle intermediates and nitric oxide levels after congenital heart surgery. Journal of Pediatrics, 2003, 142, 26-30.	1.8	59
32	Recommendations for locus-specific databases and their curation. Human Mutation, 2008, 29, 2-5.	2.5	59
33	Effective hemodialysis and hemofiltration driven by an extracorporeal membrane oxygenation pump in infants with hyperammonemia. Journal of Pediatrics, 1996, 128, 379-382.	1.8	57
34	Relationship Between Carbamoyl-Phosphate Synthetase Genotype and Systemic Vascular Function. Hypertension, 2004, 43, 186-191.	2.7	56
35	Effects of Meals High in Carbohydrate, Protein, and Fat on Ghrelin and Peptide YY Secretion in Prepubertal Children. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 4463-4471.	3.6	55
36	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55

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37	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
38	Characterization of genomic structure and polymorphisms in the human carbamyl phosphate synthetase I gene. Gene, 2003, 311, 51-57.	2.2	48
39	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. Human Mutation, 2012, 33, 165-179.	2.5	45
40	Genetic variation in the urea cycle: a model resource for investigating key candidate genes for common diseases. Human Mutation, 2009, 30, 56-60.	2.5	43
41	Nutritional Management of Urea Cycle Disorders. Critical Care Clinics, 2005, 21, S27-S35.	2.6	42
42	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
43	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
44	Evaluation of Immunization Rates and Safety Among Children With Inborn Errors of Metabolism. Pediatrics, 2011, 127, e1139-e1146.	2.1	38
45	Urinary phenylacetylglutamine as dosing biomarker for patients with urea cycle disorders. Molecular Genetics and Metabolism, 2012, 107, 308-314.	1.1	38
46	Malignancy in Noonan syndrome and related disorders. Clinical Genetics, 2015, 88, 516-522.	2.0	38
47	Development and evaluation of a machine learning-based point-of-care screening tool for genetic syndromes in children: a multinational retrospective study. The Lancet Digital Health, 2021, 3, e635-e643.	12.3	38
48	The hemochromatosis C282Y allele: a risk factor for hepatic veno-occlusive disease after hematopoietic stem cell transplantation. Bone Marrow Transplantation, 2005, 35, 1155-1164.	2.4	37
49	Late onset N-acetylglutamate synthase deficiency caused by hypomorphic alleles. Human Mutation, 2005, 25, 293-298.	2.5	37
50	Population screening in a Druze community: the challenge and the reward. Genetics in Medicine, 2008, 10, 903-909.	2.4	37
51	Linkage Relationships of Human Arginine Vasopressin-Neurophysin-II and Oxytocin-Neurophysin-I to Prodynorphin and Other Loci on Chromosome 20. Molecular Endocrinology, 1990, 4, 947-950.	3.7	36
52	Assignment of the gene (GLCLC) that encodes the heavy subunit of γ-glutamylcysteine synthetase to human chromosome 6. Cytogenetic and Genome Research, 1995, 70, 278-279.	1.1	36
53	Uniparental disomy of chromosome 2 resulting in lethal trifunctional protein deficiency due to homozygous ?-subunit mutations. Human Mutation, 2002, 20, 447-451.	2.5	36
54	L-citrulline provides a novel strategy for treating chronic pulmonary hypertension in newborn infants. Acta Paediatrica, International Journal of Paediatrics, 2014, 103, 1019-1026.	1.5	36

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55	Rapid deployment of a telemedicine care model for genetics and metabolism during <scp>COVID</scp>â€19. American Journal of Medical Genetics, Part A, 2021, 185, 68-72.	1.2	36
56	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€"A successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93-106.	3.6	35
57	Molecular Characterization of Carbamoyl-Phosphate Synthetase (CPS1) Deficiency Using Human Recombinant CPS1 as a Key Tool. Human Mutation, 2013, 34, 1149-1159.	2.5	34
58	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. Mitochondrion, 2007, 7, 204-210.	3.4	33
59	Quantitative RT-PCR comparison of the urea and nitric oxide cycle gene transcripts in adult human tissues. Molecular Genetics and Metabolism, 2009, 97, 121-127.	1.1	32
60	Vaccines Are Not Associated With Metabolic Events in Children With Urea Cycle Disorders. Pediatrics, 2011, 127, e1147-e1153.	2.1	31
61	Connecting the dots between genes, biochemistry, and disease susceptibility: systems biology modeling in human genetics. Molecular Genetics and Metabolism, 2005, 84, 104-111.	1.1	30
62	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
63	Prospective versus clinical diagnosis and therapy of acute neonatal hyperammonaemia in two sisters with carbamyl phosphate synthetase deficiency. Journal of Inherited Metabolic Disease, 1992, 15, 269-277.	3.6	26
64	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
65	Identification of dysmorphic syndromes using landmark-specific local texture descriptors. , 2016, , .		26
66	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
67	Molecular Cloning, Sequencing, Chromosomal Localization, and Tissue Distribution of the Human Na+/H+Exchanger (SLC9A2). Genomics, 1995, 30, 25-30.	2.9	23
68	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
69	Linkage analysis of the human dopamine Î²-hydroxylase gene. Genomics, 1991, 10, 493-495.	2.9	21
70	Abnormal Newborn Screens and Acylcarnitines in HIV-exposed and ARV-exposed Infants. Pediatric Infectious Disease Journal, 2013, 32, 146-150.	2.0	19
71	Hierarchical Constrained Local Model Using ICA and Its Application to Down Syndrome Detection. Lecture Notes in Computer Science, 2013, 16, 222-229.	1.3	19
72	Comparison of alterations in amino acids content in cultured astrocytes or neurons exposed to methylmercury separately or in co-culture. Neurochemistry International, 2009, 55, 136-142.	3.8	17

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73	Simple and inexpensive quantification of ammonia in whole blood. Molecular Genetics and Metabolism, 2015, 115, 95-100.	1.1	17
74	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
75	Acylcarnitine Profiles in HIV-Exposed, Uninfected Neonates in the United States. AIDS Research and Human Retroviruses, 2016, 32, 339-348.	1.1	14
76	Recurrent mutations in the vasopressin-neurophysin II gene cause autosomal dominant neurohypophyseal diabetes insipidus. Journal of Clinical Endocrinology and Metabolism, 1996, 81, 2328-2334.	3.6	14
77	Prolonged hypoxia augments l-citrulline transport by System A in the newborn piglet pulmonary circulation. Cardiovascular Research, 2012, 95, 375-384.	3.8	13
78	Down syndrome detection from facial photographs using machine learning techniques. Proceedings of SPIE, 2013, , .	0.8	13
79	Genetic susceptibility to endomyocardial fibrosis. Global Cardiology Science & Practice, 2014, 2014, 60.	0.4	12
80	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. Journal of Trauma, 2009, 66, 1265-1272.	2.3	11
81	A phase 1 doseâ€finding study of intravenous Lâ€citrulline in sickle cell disease: a potential novel therapy for sickle cell pain crisis. British Journal of Haematology, 2019, 184, 634-636.	2.5	10
82	Automated down syndrome detection using facial photographs. , 2013, 2013, 3670-3.		8
83	Ensemble learning for the detection of facial dysmorphology. , 2014, 2014, 754-7.		8
84	Combined <scp>l</scp>-citrulline and tetrahydrobiopterin therapy improves NO signaling and ameliorates chronic hypoxia-induced pulmonary hypertension in newborn pigs. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2020, 318, L762-L772.	2.9	8
85	Climbing the Ladder: Experience with Developing a Large Group Genetic Counselor Career Ladder at Childrenâ€™s National Health System. Journal of Genetic Counseling, 2016, 25, 644-648.	1.6	7
86	Metabolic basis of pediatric heart disease. Progress in Pediatric Cardiology, 2005, 20, 143-159.	0.4	6
87	Significant Differences in Markers of Oxidant Injury between Idiopathic and Bronchopulmonary-Dysplasia-Associated Pulmonary Hypertension in Children. Pulmonary Medicine, 2012, 2012, 1-6.	1.9	6
88	Diagnostic dilemma caused by overlapping features of Prader-Willi syndrome and trisomy 18 during infancy. Journal of Pediatrics, 2000, 136, 135-136.	1.8	4
89	Understanding the role of NOS-3 in ventilator-induced lung injury: don't take NO for an answer. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2010, 299, L147-L149.	2.9	4
90	Peptide tyrosine tyrosine levels are increased in patients with urea cycle disorders. Molecular Genetics and Metabolism, 2012, 106, 39-42.	1.1	4

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91	Save the biochemical geneticists!. Molecular Genetics and Metabolism, 2014, 113, 2-3.	1.1	4
92	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€"a successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93.	3.6	4
93	Mind the Gap. Molecular Genetics and Metabolism, 2014, 113, 1.	1.1	3
94	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
95	Constrained local model with independent component analysis and kernel density estimation: Application to down syndrome detection. , 2015, , .		3
96	Response to letter, broken bones, and irresponsible testimony: Enough is enough already: The flawed Ehlersâ€"Danlos syndrome infant fragility theory should not rule. American Journal of Medical Genetics, Part A, 2019, 179, 2335-2337.	1.2	3
97	Invited commentary: His life was lost but his heart still beats: In honor of children harmed by child abuse. American Journal of Medical Genetics, Part A, 2019, 179, 2329-2332.	1.2	3
98	Folic acid, either solely or combined with Lâ€citrulline, improves NO signaling and ameliorates chronic hypoxiaâ€induced pulmonary hypertension in newborn pigs. Physiological Reports, 2021, 9, e15096.	1.7	1
99	The Rare Disease Research Scholars Program: A training curriculum for clinical researchers with mixed methods evaluation study. Translational Science of Rare Diseases, 2022, 6, 1-11.	1.5	1
100	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
101	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
102	Lâ€Citrulline Inhibits Chronic Hypoxiaâ€Induced Pulmonary Hypertension in Newborn Piglets. FASEB Journal, 2008, 22, 1209.12.	0.5	0
103	A machine learning-based screening tool for genetic syndromes in children â€" Authors' reply. The Lancet Digital Health, 2022, 4, e296.	12.3	0
104	Acylcarnitines and Genetic Variation in Fat Oxidation Genes in HIV-infected, Antiretroviral-treated Children With and Without Myopathy. Pediatric Infectious Disease Journal, 2022, 41, e306-e311.	2.0	0