Bruce A Barshop

List of Publications by Year
in descending order

89 papers	5,071 citations	108046 37 h-index	100535 70 g-index
94 all docs	94 docs citations	94 times ranked	6546 citing authors

#	Article	IF	Citations
1	eP235: Interim results of the Vigilan observational study: clinical characteristics of creatine transporter deficiency. Genetics in Medicine, 2022, 24, S149.	1.1	0
2	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146
3	Tumor metabolism and neurocognition in CNS lymphoma. Neuro-Oncology, 2021, 23, 1668-1679.	0.6	9
4	Dairy Fat Intake, Plasma Pentadecanoic Acid, and Plasma Isoâ€heptadecanoic Acid Are Inversely Associated With Liver Fat in Children. Journal of Pediatric Gastroenterology and Nutrition, 2021, 72, e90-e96.	0.9	16
5	A post glycosylphosphatidylinositol (GPI) attachment to proteins, type 2 (PGAP2) variant identified in Mabry syndrome index cases: Molecular genetics of the prototypical inherited GPI disorder. European Journal of Medical Genetics, 2020, 63, 103822.	0.7	8
6	Cardiac tissue citric acid cycle intermediates in exercised very longâ€chain <scp>acylâ€CoA</scp> dehydrogenaseâ€deficient mice fed triheptanoin or mediumâ€chain triglyceride. Journal of Inherited Metabolic Disease, 2020, 43, 1232-1242.	1.7	6
7	Identification of pathognomonic purine synthesis biomarkers by metabolomic profiling of adolescents with obesity and type 2 diabetes. PLoS ONE, 2020, 15, e0234970.	1.1	21
8	Cystathionine beta synthase deficiency and brain edema associated with methionine excess under betaine supplementation: Four new cases and a review of the evidence. JIMD Reports, 2020, 52, 3-10.	0.7	12
9	Thiamine phosphokinase deficiency and mutation in TPK1 presenting as biotin responsive basal ganglia disease. Clinica Chimica Acta, 2019, 499, 13-15.	0.5	8
10	Promises and pitfalls of untargeted metabolomics. Journal of Inherited Metabolic Disease, 2018, 41, 355-366.	1.7	149
11	Brain Magnetic Resonance Imaging Findings in Poorly Controlled Homocystinuria. Journal of Radiology Case Reports, 2018, 12, 1-8.	0.2	4
12	Biallelic loss of human CTNNA2, encoding Î±N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. Nature Genetics, 2018, 50, 1093-1101.	9.4	70
13	The mPEG-PCL Copolymer for Selective Fermentation of Staphylococcus lugdunensis Against Candida parapsilosis in the Human Microbiome. Journal of Microbial & Biochemical Technology, 2016, 8, 259-265.	0.2	6
14	Controversies and research agenda in nephropathic cystinosis: conclusions from a â€œKidney Disease: Improving Global Outcomesâ€•(KDIGO) Controversies Conference. Kidney International, 2016, 89, 1192-1203.	2.6	52
15	Paracentric Inversion of Chromosome 21 Leading to Disruption of the HLCS Gene in a Family with Holocarboxylase Synthetase Deficiency. JIMD Reports, 2016, 34, 55-61.	0.7	1
16	Diagnosis and Monitoring of Cystinosis Using Immunomagnetically Purified Granulocytes. Clinical Chemistry, 2016, 62, 766-772.	1.5	13
17	p300 is not required for metabolic adaptation to endurance exercise training. FASEB Journal, 2016, 30, 1623-1633.	0.2	21
18	In vivo monitoring of urea cycle activity with 13C-acetate as a tracer of ureagenesis. Molecular Genetics and Metabolism, 2016, 117, 19-26.	0.5	5

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19	Perturbations of tyrosine metabolism promote the indolepyruvate pathway via tryptophan in host and microbiome. Molecular Genetics and Metabolism, 2015, 114, 431-437.	0.5	39
20	Consanguinity and rare mutations outside of MCCC genes underlie nonspecific phenotypes of MCCD. Genetics in Medicine, 2015, 17, 660-667.	1.1	9
21	Metabolic Effects of Increasing Doses of Nitisinone in the Treatment of Alkaptonuria. JIMD Reports, 2015, 24, 13-20.	0.7	16
22	Pharmacokinetics of cysteamine bitartrate following intraduodenal delivery. Fundamental and Clinical Pharmacology, 2014, 28, 136-143.	1.0	14
23	Infants suspected to have very-long chain acyl-CoA dehydrogenase deficiency from newborn screening. Molecular Genetics and Metabolism, 2014, 111, 484-492.	0.5	47
24	A modelâ€driven quantitative metabolomics analysis of aerobic and anaerobic metabolism in E. coli Kâ€12 MG1655 that is biochemically and thermodynamically consistent. Biotechnology and Bioengineering, 2014, 111, 803-815.	1.7	53
25	Validation of a dual LCâ€“HRMS platform for clinical metabolic diagnosis in serum, bridging quantitative analysis and untargeted metabolomics. Metabolomics, 2014, 10, 312-323.	1.4	59
26	Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring. Molecular Genetics and Metabolism, 2014, 111, 16-25.	0.5	111
27	Cysteamine Modulates Oxidative Stress and Blocks Myofibroblast Activity in CKD. Journal of the American Society of Nephrology: JASN, 2014, 25, 43-54.	3.0	58
28	Elevation of guanidinoacetate in newborn dried blood spots and impact of early treatment in GAMT deficiency. Molecular Genetics and Metabolism, 2013, 109, 215-217.	0.5	38
29	Metabolomics Reveals Signature of Mitochondrial Dysfunction in Diabetic Kidney Disease. Journal of the American Society of Nephrology: JASN, 2013, 24, 1901-1912.	3.0	454
30	Changes in plasma and urine globotriaosylceramide levels do not predict Fabry disease progression over 1 year of agalsidase alfa. Genetics in Medicine, 2013, 15, 983-989.	1.1	21
31	Biochemical, molecular, and clinical characteristics of children with short chain acyl-CoA dehydrogenase deficiency detected by newborn screening in California. Molecular Genetics and Metabolism, 2012, 106, 55-61.	0.5	65
32	Homocystinuria and Hyperhomocysteinemia. , 2012, , 1361-1363.		1
33	Exome sequencing identifies ACSF3 as a cause of combined malonic and methylmalonic aciduria. Nature Genetics, 2011, 43, 883-886.	9.4	89
34	Novel mutations in the human MCCA and MCCB gene causing methylcrotonylglycinuria. Molecular Genetics and Metabolism, 2011, 102, 218-221.	0.5	10
35	45-Year-old female with propionic acidemia, renal failure, and premature ovarian failure; late complications of propionic acidemia?. Molecular Genetics and Metabolism, 2011, 103, 338-340.	0.5	27
36	Twice-Daily Cysteamine Bitartrate Therapy for Children with Cystinosis. Journal of Pediatrics, 2010, 156, 71-75.e3.	0.9	48

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37	Long-Term Treatment of Cystinosis in Children with Twice-Daily Cysteamine. Journal of Pediatrics, 2010, 156, 823-827.	0.9	27
38	Pharmacokinetics of enteric-coated cysteamine bitartrate in healthy adults: a pilot study. British Journal of Clinical Pharmacology, 2010, 70, 376-382.	1.1	22
39	Agalsidase Alfa and Kidney Dysfunction in Fabry Disease. Journal of the American Society of Nephrology: JASN, 2009, 20, 1132-1139.	3.0	148
40	Time before isolating cystinotic leukocytes affects reliability of cystine determination. Pediatric Nephrology, 2009, 24, 2465-2466.	0.9	1
41	Expanded clinical and molecular spectrum of guanidinoacetate methyltransferase (GAMT) deficiency. Molecular Genetics and Metabolism, 2009, 96, 38-43.	0.5	70
42	Laboratory referral practices in biochemical genetics in the United States. Molecular Genetics and Metabolism, 2009, 98, 149-151.	0.5	0
43	A Delphi-based consensus clinical practice protocol for the diagnosis and management of 3-methylcrotonyl CoA carboxylase deficiency. Molecular Genetics and Metabolism, 2008, 93, 363-370.	0.5	58
44	Attitudes regarding vaccination among practitioners of clinical biochemical genetics. Molecular Genetics and Metabolism, 2008, 95, 1-2.	0.5	4
45	Management of a patient with holocarboxylase synthetase deficiency. Molecular Genetics and Metabolism, 2008, 95, 201-205.	0.5	39
46	Multiple organic anion transporters contribute to net renal excretion of uric acid. Physiological Genomics, 2008, 33, 180-192.	1.0	203
47	Organic Anion Transporter 3 Contributes to the Regulation of Blood Pressure. Journal of the American Society of Nephrology: JASN, 2008, 19, 1732-1740.	3.0	72
48	Potential Misdiagnosis of 3-Methylcrotonyl-Coenzyme A Carboxylase Deficiency Associated With Absent or Trace Urinary 3-Methylcrotonylglycine. Pediatrics, 2007, 120, e1335-e1340.	1.0	17
49	Fatal Initial Adult-Onset Presentation of Urea Cycle Defect. Archives of Neurology, 2007, 64, 1777.	4.9	37
50	Analysis of coenzyme Q in human blood and tissues. Mitochondrion, 2007, 7, S89-S93.	1.6	53
51	Metabolomics Identifies Perturbations in Human Disorders of Propionate Metabolism. Clinical Chemistry, 2007, 53, 2169-2176.	1.5	148
52	Vomitingâ€”Again?. Journal of Hospital Medicine, 2007, 2, 189-193.	0.7	0
53	Pharmacokinetics of cysteamine bitartrate following gastrointestinal infusion. British Journal of Clinical Pharmacology, 2007, 63, 36-40.	1.1	26
54	Understanding intestinal cysteamine bitartrate absorption. Journal of Pediatrics, 2006, 148, 764-769.	0.9	38

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55	Domino liver transplantation in maple syrup urine disease. Liver Transplantation, 2006, 12, 876-882.	1.3	69
56	Decreased Renal Organic Anion Secretion and Plasma Accumulation of Endogenous Organic Anions in OAT1 Knock-out Mice. Journal of Biological Chemistry, 2006, 281, 5072-5083.	1.6	204
57	Disorders of Valine-Isoleucine Metabolism. , 2006, , 81-92.		0
58	Biotinidase deficiency: Novel mutations and their biochemical and clinical correlates. Human Mutation, 2005, 25, 413-413.	1.1	28
59	Esomeprazole therapy for gastric acid hypersecretion in children with cystinosis. Pediatric Nephrology, 2005, 20, 1786-1793.	0.9	42
60	Domino Hepatic Transplantation in Maple Syrup Urine Disease. New England Journal of Medicine, 2005, 353, 2410-2411.	13.9	37
61	The Role of Methionine in Ethylmalonic Encephalopathy with Petechiae. Archives of Neurology, 2004, 61, 570.	4.9	19
62	Chronic treatment of mitochondrial disease patients with dichloroacetate. Molecular Genetics and Metabolism, 2004, 83, 138-149.	0.5	53
63	Metabolomic approaches to mitochondrial disease: correlation of urine organic acids. Mitochondrion, 2004, 4, 521-527.	1.6	64
64	Total plasma homocysteine and primary open-angle glaucoma. American Journal of Ophthalmology, 2004, 137, 401-406.	1.7	24
65	Identification of 16 novel mutations in the argininosuccinate synthetase gene and genotype-phenotype correlation in 38 classical citrullinemia patients. Human Mutation, 2003, 22, 24-34.	1.1	71
66	Effects of estrogen and psychological stress on plasma homocysteine levels. Fertility and Sterility, 2003, 79, 256-260.	0.5	20
67	Fructo-oligosaccharide tolerance in patients with hereditary fructose intolerance. A preliminary nonrandomized open challenge short-term study. Nutrition Research, 2003, 23, 1003-1011.	1.3	8
68	Role of L-Carnitine in Apnea of Prematurity: A Randomized, Controlled Trial. Pediatrics, 2002, 109, 622-626.	1.0	19
69	Pyruvate carboxylase deficiencyâ€"insights from liver transplantation. Molecular Genetics and Metabolism, 2002, 77, 143-149.	0.5	25
70	Mitochondrial tubulopathy: the many faces of mitochondrial disorders. Pediatric Nephrology, 2001, 16, 710-712.	0.9	25
71	Creatine kinase and uric acid: early warning for metabolic imbalance resulting from disorders of fatty acid oxidation. European Journal of Pediatrics, 2001, 160, 599-602.	1.3	20
72	Nerve conduction changes in patients with mitochondrial diseases treated with dichloroacetate. Muscle and Nerve, 2001, 24, 916-924.	1.0	55

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73	Autism Associated With the Mitochondrial DNA G8363A Transfer RNALys Mutation. Journal of Child Neurology, 2000, 15, 357-361.	0.7	153
74	Kearnsâ€“Sayre Syndrome Presenting as 2-Oxoadipic Aciduria. Molecular Genetics and Metabolism, 2000, 69, 64-68.	0.5	14
75	Mitochondrial DNA polymerase ? deficiency and mtDNA depletion in a child with Alpers' syndrome. Annals of Neurology, 1999, 45, 54-58.	2.8	192
76	Diet change in the management of metabolic encephalomyopathies. BioFactors, 1998, 7, 259-262.	2.6	3
77	Analysis of organic acids, amino acids, and carnitine in dogs with lipid storage myopathy. , 1998, 21, 1202-1205.		22
78	Oxidative Metabolism in Rett Syndrome: 1. Clinical Studies. Neuropediatrics, 1995, 26, 90-94.	0.3	21
79	A mutation in adenylosuccinate lyase associated with mental retardation and autistic features. Nature Genetics, 1992, 1, 59-63.	9.4	127
80	Luminescent immobilized enzyme test systems for inorganic pyrophosphate: Assays using firefly luciferase and nicotinamide-mononucleotide adenylyl transferase or adenosine-5â€²-triphosphate sulfurylase. Analytical Biochemistry, 1991, 197, 266-272.	1.1	45
81	Metabolism of 1-13C-Propionate In Vivo in Patients with Disorders of Propionate Metabolism. Pediatric Research, 1991, 30, 15-22.	1.1	27
82	3-Hydroxyisobutyric Aciduria: An Inborn Error of Valine Metabolism. Pediatric Research, 1991, 30, 322-326.	1.1	33
83	Metabolism of 1-13C-Propionate In Vivo in Patients with Disorders of Propionate Metabolism. Pediatric Research, 1991, 30, 15???22.	1.1	0
84	Transcobalamin II deficiency presenting with methylmalonic aciduria and homocystinuria and abnormal absorption of cobalamin. American Journal of Medical Genetics Part A, 1990, 35, 222-228.	2.4	34
85	Effects of Ascorbic Acid in Alkaptonuria: Alterations in Benzoquinone Acetic Acid and an Ontogenic Effect in Infancy. Pediatric Research, 1989, 26, 140-144.	1.1	99
86	Duplication of 16q and deletion of 15q. American Journal of Medical Genetics Part A, 1989, 34, 183-186.	2.4	20
87	Analysis of numerical methods for computer simulation of kinetic processes: Development of KINSIMâ€”A flexible, portable system. Analytical Biochemistry, 1983, 130, 134-145.	1.1	705
88	Effect of DTNB light chain on the interaction of vertebrate skeletal myosin with actin. Nature, 1975, 258, 163-166.	13.7	114
89	Atlas of Inherited Metabolic Diseases 3E. , 0, , .		8