Gerard Berry

List of Publications by Year
in descending order

178 papers	7,597 citations	47006 47 h-index	66911 78 g-index
188 all docs	188 docs citations	188 times ranked	8639 citing authors

#	Article	IF	Citations
1	Survival after Treatment with Phenylacetate and Benzoate for Urea-Cycle Disorders. New England Journal of Medicine, 2007, 356, 2282-2292.	27.0	320
2	Deletions of NRXN1 (neurexinâ€1) predispose to a wide spectrum of developmental disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 937-947.	1.7	217
3	A Deficiency of Carnitineâ€“Acylcarnitine Translocase in the Inner Mitochondrial Membrane. New England Journal of Medicine, 1992, 327, 19-23.	27.0	183
4	Uremic solutes and risk of end-stage renal disease in type 2 diabetes: metabolomic study. Kidney International, 2014, 85, 1214-1224.	5.2	182
5	Galactose metabolism and health. Current Opinion in Clinical Nutrition and Metabolic Care, 2015, 18, 422-427.	2.5	162
6	MR diffusion imaging and MR spectroscopy of maple syrup urine disease during acute metabolic decompensation. Neuroradiology, 2003, 45, 393-399.	2.2	157
7	Transcription of the Sodium/myo-Inositol Cotransporter Gene Is Regulated by Multiple Tonicity-responsive Enhancers Spread over 50 Kilobase Pairs in the 5â€²-Flanking Region. Journal of Biological Chemistry, 1998, 273, 20615-20621.	3.4	155
8	Targeted exome sequencing of suspected mitochondrial disorders. Neurology, 2013, 80, 1762-1770.	1.1	155
9	The adult galactosemic phenotype. Journal of Inherited Metabolic Disease, 2012, 35, 279-286.	3.6	151
10	Exploring concordance and discordance for return of incidental findings from clinical sequencing. Genetics in Medicine, 2012, 14, 405-410.	2.4	149
11	The mutation spectrum of the facilitative glucose transporter gene SLC2A2 (GLUT2) in patients with Fanconi-Bickel syndrome. Human Genetics, 2002, 110, 21-29.	3.8	147
12	Copy number variation plays an important role in clinical epilepsy. Annals of Neurology, 2014, 75, 943-958.	5.3	147
13	Acute extrapyramidal syndrome in methylmalonic acidemia: â€œMetabolic strokeâ€•involving the globus pallidus. Journal of Pediatrics, 1988, 113, 1022-1027.	1.8	135
14	International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and followâ€up. Journal of Inherited Metabolic Disease, 2017, 40, 171-176.	3.6	132
15	The human osmoregulatory Na+/myo-inositol cotransporter gene (SLC5A3): molecular cloning and localization to chromosome 21. Genomics, 1995, 25, 507-513.	2.9	119
16	Bumetanide Enhances Phenobarbital Efficacy in a Rat Model of Hypoxic Neonatal Seizures. PLoS ONE, 2013, 8, e57148.	2.5	117
17	Cytochrome c oxidase-associated Leigh syndrome: Phenotypic features and pathogenetic speculations. Journal of the Neurological Sciences, 1991, 104, 97-111.	0.6	108
18	Homozygous PLCB1 deletion associated with malignant migrating partial seizures in infancy. Epilepsia, 2012, 53, e146-50.	5.1	104

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19	A syndrome of congenital hyperinsulinism and hyperammonemia. Journal of Pediatrics, 1997, 130, 661-664.	1.8	101
20	Risk factors for premature ovarian failure in females with galactosemia. Journal of Pediatrics, 2000, 137, 833-841.	1.8	99
21	Targeting proximal tubule mitochondrial dysfunction attenuates the renal disease of methylmalonic acidemia. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 13552-13557.	7.1	97
22	Glycogen synthase kinase-3 is essential for Î²-arrestin-2 complex formation and lithium-sensitive behaviors in mice. Journal of Clinical Investigation, 2011, 121, 3756-3762.	8.2	96
23	Newborn Screening by Tandem Mass Spectrometry for Medium-Chain Acyl-CoA Dehydrogenase Deficiency: A Cost-Effectiveness Analysis. Pediatrics, 2003, 112, 1005-1015.	2.1	90
24	High-performance liquid chromatography of phospholipids with UV detection: optimization of separations on silica. Biomedical Applications, 1981, 225, 319-328.	1.7	86
25	Hereditary galactosemia. Metabolism: Clinical and Experimental, 2018, 83, 188-196.	3.4	82
26	Intellectual outcome in children with maple syrup urine disease. Journal of Pediatrics, 1991, 119, 46-50.	1.8	81
27	Loss of Murine Na+/myo-Inositol Cotransporter Leads to Brain myo-Inositol Depletion and Central Apnea. Journal of Biological Chemistry, 2003, 278, 18297-18302.	3.4	80
28	Epimerase-Deficiency Galactosemia Is Not a Binary Condition. American Journal of Human Genetics, 2006, 78, 89-102.	6.2	77
29	Diversity of approaches to classic galactosemia around the world: a comparison of diagnosis, intervention, and outcomes. Journal of Inherited Metabolic Disease, 2012, 35, 1037-1049.	3.6	77
30	Fatal Hyperammonemia after Orthotopic Lung Transplantation. Annals of Internal Medicine, 2000, 132, 283.	3.9	76
31	The rate of de novo galactose synthesis in patients with galactose-1-phosphate uridyltransferase deficiency. Molecular Genetics and Metabolism, 2004, 81, 22-30.	1.1	75
32	Liver transplantation is not curative for methylmalonic acidopathy caused by methylmalonyl-CoA mutase deficiency. Molecular Genetics and Metabolism, 2006, 88, 322-326.	1.1	72
33	Disorders of Galactose Metabolism. , 2006, , 121-130.		69
34	In vivo evidence of brain galactitol accumulation in an infant with galactosemia and encephalopathy. Journal of Pediatrics, 2001, 138, 260-262.	1.8	68
35	Galactosemia: When is it a newborn screening emergency?. Molecular Genetics and Metabolism, 2012, 106, 7-11.	1.1	68
36	Sodium-myoinositol cotransporter-1, SMIT1, mediates the production of reactive oxygen species induced by hyperglycemia in the heart. Scientific Reports, 2017, 7, 41166.	3.3	64

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37	Long-term management of patients with urea cycle disorders. Journal of Pediatrics, 2001, 138, S56-S61.	1.8	62
38	The role of polyols in the pathophysiology of hypergalactosemia. European Journal of Pediatrics, 1995, 154, S53-S64.	2.7	61
39	Short-chain acyl-CoA dehydrogenase gene mutation (c.319C>T) presents with clinical heterogeneity and is candidate founder mutation in individuals of Ashkenazi Jewish origin. Molecular Genetics and Metabolism, 2008, 93, 179-189.	1.1	61
40	DDOST Mutations Identified by Whole-Exome Sequencing Are Implicated in Congenital Disorders of Glycosylation. American Journal of Human Genetics, 2012, 90, 363-368.	6.2	60
41	Branched-Chain Amino Acid-Free Parenteral Nutrition in the Treatment of Acute Metabolic Decompensation in Patients with Maple Syrup Urine Disease. New England Journal of Medicine, 1991, 324, 175-179.	27.0	56
42	KCNQ1, KCNE2, and Na ⁺ -Coupled Solute Transporters Form Reciprocally Regulating Complexes That Affect Neuronal Excitability. Science Signaling, 2014, 7, ra22.	3.6	56
43	Galactose Metabolism by the Mouse with Galactose-1-Phosphate Uridyltransferase Deficiency. Pediatric Research, 2000, 48, 211-217.	2.3	54
44	Urinary Galactonate in Patients with Galactosemia: Quantitation by Nuclear Magnetic Resonance Spectroscopy. Pediatric Research, 1997, 42, 855-861.	2.3	52
45	Methylmalonic acidemia: A megamitochondrial disorder affecting the kidney. Pediatric Nephrology, 2014, 29, 2139-2146.	1.7	52
46	Phosphoinositide deficiency due to inositol depletion is not a mechanism of lithium action in brain. Molecular Genetics and Metabolism, 2004, 82, 87-92.	1.1	51
47	Whole genome sequencing identifies <scp>SCN2A</scp> mutation in monozygotic twins with <scp>O</scp>htahara syndrome and unique neuropathologic findings. Epilepsia, 2013, 54, e81-5.	5.1	49
48	A re-evaluation of life-long severe galactose restriction for the nutrition management of classic galactosemia. Molecular Genetics and Metabolism, 2014, 112, 191-197.	1.1	49
49	Knockout mice in understanding the mechanism of action of lithium. Biochemical Society Transactions, 2009, 37, 1121-1125.	3.4	48
50	Succinyl-CoA:3-Ketoacid CoA Transferase (SCOT): Cloning of the Human SCOT Gene, Tertiary Structural Modeling of the Human SCOT Monomer, and Characterization of Three Pathogenic Mutations. Genomics, 2000, 68, 144-151.	2.9	47
51	Results from a 78â€week, singleâ€arm, openâ€label phase 2 study to evaluate UX007 in pediatric and adult patients with severe longâ€chain fatty acid oxidation disorders (LCâ€FAOD). Journal of Inherited Metabolic Disease, 2019, 42, 169-177.	3.6	47
52	Is prenatal myoâ€inositol deficiency a mechanism of CNS injury in galactosemia?. Journal of Inherited Metabolic Disease, 2011, 34, 345-355.	3.6	46
53	A novel de novo mutation inATP1A3and childhood-onset schizophrenia. Journal of Physical Education and Sports Management, 2016, 2, a001008.	1.2	46
54	Isovaleric acidemia: Medical and neurodevelopmental effects of long-term therapy. Journal of Pediatrics, 1988, 113, 58-64.	1.8	45

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55	Urine and plasma galactitol in patients with galactose-1-phosphate uridyltransferase deficiency galactosemia. Metabolism: Clinical and Experimental, 1999, 48, 1294-1302.	3.4	45
56	Galactose Breath Testing Distinguishes Variant and Severe Galactose-1-Phosphate Uridyltransferase Genotypes. Pediatric Research, 2000, 48, 323-328.	2.3	44
57	Sensitive isotope dilution liquid chromatography/tandem mass spectrometry method for quantitative analysis of bumetanide in serum and brain tissue. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2011, 879, 998-1002.	2.3	44
58	Diabetes-like renal glomerular disease in Fanconi-Bickel syndrome. Pediatric Nephrology, 1995, 9, 287-291.	1.7	43
59	Characterization of the null murine sodium/myo-inositol cotransporter 1 (Smit1 or Slc5a3) phenotype: Myo-inositol rescue is independent of expression of its cognate mitochondrial ribosomal protein subunit 6 (Mrps6) gene and of phosphatidylinositol levels in neonatal brain. Molecular Genetics and Metabolism, 2008, 95, 81-95.	1.1	43
60	The concentration of red blood cell UDPGlucose and UDPGalactose determined by high-performance liquid chromatography. Analytical Biochemistry, 1991, 194, 388-393.	2.4	42
61	Inositol-Related Gene Knockouts Mimic Lithiumâ€™s Effect on Mitochondrial Function. Neuropsychopharmacology, 2014, 39, 319-328.	5.4	42
62	KCTD7 deficiency defines a distinct neurodegenerative disorder with a conserved autophagyâ€lysosome defect. Annals of Neurology, 2018, 84, 766-780.	5.3	42
63	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
64	Fertility in adult women with classic galactosemia and primary ovarian insufficiency. Fertility and Sterility, 2017, 108, 168-174.	1.0	42
65	Renal Handling of Carnitine in Secondary Carnitine Deficiency Disorders. Pediatric Research, 1993, 34, 89-96.	2.3	40
66	Proton magnetic resonance spectroscopy of brain metabolites in galactosemia. Annals of Neurology, 2001, 50, 266-269.	5.3	40
67	Introduction to the Maastricht workshop: lessons from the past and new directions in galactosemia. Journal of Inherited Metabolic Disease, 2011, 34, 249-255.	3.6	40
68	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	2.5	40
69	The Structural Organization of the Human Na+/Myo-inositol Cotransporter (SLC5A3) Gene and Characterization of the Promoter. Genomics, 1997, 46, 459-465.	2.9	39
70	In vivo brain myo -inositol levels in children with Down syndrome. Journal of Pediatrics, 1999, 135, 94-97.	1.8	39
71	In vivo pyruvate detected by MR spectroscopy in neonatal pyruvate dehydrogenase deficiency. American Journal of Neuroradiology, 2003, 24, 1471-4.	2.4	39
72	Evidence for Alternate Galactose Oxidation in a Patient with Deletion of the Galactose-1-Phosphate Uridyltransferase Gene. Molecular Genetics and Metabolism, 2001, 72, 316-321.	1.1	38

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73	A compound heterozygous mutation in GPD1 causes hepatomegaly, steatohepatitis, and hypertriglyceridemia. European Journal of Human Genetics, 2014, 22, 1229-1232.	2.8	38
74	Novel founder intronic variant in SLC39A14 in two families causing Manganism and potential treatment strategies. Molecular Genetics and Metabolism, 2018, 124, 161-167.	1.1	36
75	Effects of triheptanoin (<scp>UX007</scp>) in patients with longâ€chain fatty acid oxidation disorders: Results from an <scp>openâ€label</scp>, <scp>longâ€term</scp> extension study. Journal of Inherited Metabolic Disease, 2021, 44, 253-263.	3.6	36
76	Phosphatidylcholine removal from brain lipid extracts expands lipid detection and enhances phosphoinositide quantification by matrix-assisted laser desorption/ionization time-of-flight (MALDI-TOF) mass spectrometry. Analytical Biochemistry, 2007, 362, 155-167.	2.4	34
77	Galactosemia and Amenorrhea in the Adolescent. Annals of the New York Academy of Sciences, 2008, 1135, 112-117.	3.8	34
78	N- and O-linked glycosylation of total plasma glycoproteins in galactosemia. Molecular Genetics and Metabolism, 2012, 106, 442-454.	1.1	34
79	Fertility preservation in female classic galactosemia patients. Orphanet Journal of Rare Diseases, 2013, 8, 107.	2.7	34
80	Red blood cell uridine sugar nucleotide levels in patients with classic galactosemia and other metabolic disorders. Metabolism: Clinical and Experimental, 1992, 41, 783-787.	3.4	32
81	SMIT1 haploinsufficiency causes brain inositol deficiency without affecting lithium-sensitive behavior. Molecular Genetics and Metabolism, 2006, 88, 384-388.	1.1	32
82	Galactose Metabolism in Mice with Galactose-1-Phosphate Uridyltransferase Deficiency: Sucklings and 7-Week-Old Animals Fed a High-Galactose Diet. Molecular Genetics and Metabolism, 2001, 72, 306-315.	1.1	30
83	Homozygote inositol transporter knockout mice show a lithiumâ€like phenotype. Bipolar Disorders, 2008, 10, 453-459.	1.9	29
84	Urine oligosaccharide screening by MALDI-TOF for the identification of NGLY1 deficiency. Molecular Genetics and Metabolism, 2018, 124, 82-86.	1.1	29
85	Galactose-1 phosphate uridylyltransferase (GalT) gene: A novel positive regulator of the PI3K/Akt signaling pathway in mouse fibroblasts. Biochemical and Biophysical Research Communications, 2016, 470, 205-212.	2.1	28
86	Phenotypic expansion of <scp>Boschâ€“Boonstraâ€“Schaaf</scp> optic atrophy syndrome and further evidence for genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2020, 182, 1426-1437.	1.2	27
87	31P NMR analysis of red blood cell UDPGlucose and UDPGalactose: Comparison with HPLC and enzymatic methods. Analytical Biochemistry, 1992, 202, 105-110.	2.4	26
88	Inborn Errors of Metabolism with Hepatopathy. Pediatric Clinics of North America, 2018, 65, 337-352.	1.8	26
89	Gainâ€ofâ€function variants in the ODC1 gene cause a syndromic neurodevelopmental disorder associated with macrocephaly, alopecia, dysmorphic features, and neuroimaging abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 2554-2560.	1.2	26
90	Pathophysiology and targets for treatment in hereditary galactosemia: A systematic review of animal and cellular models. Journal of Inherited Metabolic Disease, 2020, 43, 392-408.	3.6	25

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91	The male reproductive system in classic galactosemia: cryptorchidism and low semen volume. Journal of Inherited Metabolic Disease, 2013, 36, 779-786.	3.6	24
92	Succinyl-CoA synthetase (SUCLA2) deficiency in two siblings with impaired activity of other mitochondrial oxidative enzymes in skeletal muscle without mitochondrial DNA depletion. Molecular Genetics and Metabolism, 2017, 120, 213-222.	1.1	24
93	High-performance liquid chromatography of phospholipids: Quantitation by phosphate analysis. Analytical Biochemistry, 1983, 135, 239-243.	2.4	23
94	Quantification of Galactose-1-Phosphate Uridyltransferase Enzyme Activity by Liquid Chromatographyâ€“Tandem Mass Spectrometry. Clinical Chemistry, 2010, 56, 772-780.	3.2	22
95	Psychosocial developmental milestones in men with classic galactosemia. Journal of Inherited Metabolic Disease, 2011, 34, 415-419.	3.6	22
96	Use of citrulline as a diagnostic marker in the prospective treatment of urea cycle disorders. Journal of Pediatrics, 1991, 118, 914-917.	1.8	21
97	Comparison of erythrocyte uridine sugar nucleotide levels in normals, classic galactosemics, and patients with other metabolic disorders. Metabolism: Clinical and Experimental, 1995, 44, 597-604.	3.4	21
98	Galactose Content of Legumes, Caseinates, and Some Hard Cheeses: Implications for Diet Treatment of Classic Galactosemia. Journal of Agricultural and Food Chemistry, 2014, 62, 1397-1402.	5.2	21
99	Extended [13C]galactose oxidation studies in patients with galactosemia. Molecular Genetics and Metabolism, 2004, 82, 130-136.	1.1	20
100	Ultra fast and sensitive liquid chromatography tandem mass spectrometry based assay for galactose-1-phosphate uridylyltransferase and galactokinase deficiencies. Molecular Genetics and Metabolism, 2011, 102, 33-40.	1.1	20
101	Genetic Determinants of Sudden Unexpected Death in Pediatrics. Genetics in Medicine, 2022, 24, 839-850.	2.4	20
102	Kinetic evidence for compartmentalization of myo-inositol in hepatocytes. Metabolism: Clinical and Experimental, 1993, 42, 395-401.	3.4	19
103	Novel variants in SPTAN1 without epilepsy: An expansion of the phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 2768-2776.	1.2	19
104	Defining a new immune deficiency syndrome: MAN2B2-CDG. Journal of Allergy and Clinical Immunology, 2020, 145, 1008-1011.	2.9	19
105	CMP-dependent phosphatidylinositol:myo-inositol exchange activity in isolated nerve-endings. Biochemical and Biophysical Research Communications, 1983, 112, 817-821.	2.1	18
106	Rhabdomyolysis, acute renal failure, and cardiac arrest secondary to status dystonicus in a child with glutaric aciduria type I. Molecular Genetics and Metabolism, 2012, 106, 488-490.	1.1	18
107	5,10-methenyltetrahydrofolate synthetase deficiency causes a neurometabolic disorder associated with microcephaly, epilepsy, and cerebral hypomyelination. Molecular Genetics and Metabolism, 2018, 125, 118-126.	1.1	18
108	Elements of diabetic nephropathy in a patient with GLUT2 deficiency. Molecular Genetics and Metabolism, 2005, 86, 473-477.	1.1	17

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109	Witnessed sleep-related seizure and sudden unexpected death in infancy: a case report. Forensic Science, Medicine, and Pathology, 2013, 9, 418-421.	1.4	17
110	Management of a Woman With Maple Syrup Urine Disease During Pregnancy, Delivery, and Lactation. Journal of Parenteral and Enteral Nutrition, 2015, 39, 875-879.	2.6	17
111	Acute Illness Protocol for Organic Acidemias. Pediatric Emergency Care, 2017, 33, 142-146.	0.9	17
112	Phosphatidylinositol:myo-Inositol Exchange Activity in Intact Nerve Endings: Substrate and Cofactor Dependence, Nucleotide Specificity, and Effect on Synaptosomal Handling of myo-Inositol. Journal of Neurochemistry, 1986, 46, 1073-1080.	3.9	16
113	N-Acetylcysteine Therapy in an Infant with Transaldolase Deficiency Is Well Tolerated and Associated with Normalization of Alpha Fetoprotein Levels. JIMD Reports, 2016, 31, 73-77.	1.5	16
114	Impaired fertility and motor function in a zebrafish model for classic galactosemia. Journal of Inherited Metabolic Disease, 2018, 41, 117-127.	3.6	16
115	Apparent Galactose Appearance Rate in Human Galactosemia Based on Plasma [13C]Galactose Isotopic Enrichment. Molecular Genetics and Metabolism, 2000, 70, 261-271.	1.1	15
116	De Novo TUBB2A Variant Presenting With Anterior Temporal Pachygyria. Journal of Child Neurology, 2017, 32, 127-131.	1.4	15
117	Disorders of Galactose Metabolism. , 2012, , 141-150.		14
118	Untargeted metabolomics as an unbiased approach to the diagnosis of inborn errors of metabolism of the non-oxidative branch of the pentose phosphate pathway. Molecular Genetics and Metabolism, 2020, 131, 147-154.	1.1	14
119	An emerging role for endothelial barrier support therapy for congenital disorders of glycosylation. Journal of Inherited Metabolic Disease, 2020, 43, 880-890.	3.6	14
120	Galactokinase deficiency: lessons from the GalNet registry. Genetics in Medicine, 2021, 23, 202-210.	2.4	14
121	Abnormal myo-inositol and phospholipid metabolism in cultured fibroblasts from patients with ataxia telangiectasia. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 1999, 1437, 287-300.	2.4	12
122	Ornithine transcarbamylase deficiency and pancreatitis. Journal of Pediatrics, 2001, 138, 123-124.	1.8	12
123	De novo ATP1A3 and compound heterozygous NLRP3 mutations in a child with autism spectrum disorder, episodic fatigue and somnolence, and muckle-wells syndrome. Molecular Genetics and Metabolism Reports, 2018, 16, 23-29.	1.1	12
124	The reâ€•occurrence of cardiomyopathy in propionic acidemia after liver transplantation. JIMD Reports, 2020, 54, 3-8.	1.5	12
125	A retrospective study of adult patients with noncirrhotic hyperammonemia. Journal of Inherited Metabolic Disease, 2020, 43, 1165-1172.	3.6	12
126	Pathophysiology of long-term complications in classic galactosemia: What we do and do not know. Molecular Genetics and Metabolism, 2022, 137, 33-39.	1.1	12

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127	Uridine Diphosphate Hexoses in Leukocytes and Fibroblasts of Classic Galactosemics and Patients with Other Metabolic Diseases. Pediatric Research, 1994, 36, 613-618.	2.3	11
128	Disease Heterogeneity in Na+/Citrate Cotransporter Deficiency. JIMD Reports, 2016, 31, 107-111.	1.5	11
129	The ability of an LC-MS/MS-based erythrocyte GALT enzyme assay to predict the phenotype in subjects with GALT deficiency. Molecular Genetics and Metabolism, 2019, 126, 368-376.	1.1	11
130	Experimental Galactose Toxicity: Effects on Synaptosomal Phosphatidylinositol Metabolism. Journal of Neurochemistry, 1981, 37, 888-891.	3.9	10
131	Uridine diphosphate glucose and uridine diphosphate galactose in galactosemia. Journal of Pediatrics, 1990, 117, 838-840.	1.8	10
132	Recurrent unexplained hyperammonemia in an adolescent with arginase deficiency. Clinical Biochemistry, 2012, 45, 1583-1586.	1.9	10
133	A novel null mutation in the pyruvate dehydrogenase phosphatase catalytic subunit gene (PDP1) causing pyruvate dehydrogenase complex deficiency. JIMD Reports, 2019, 48, 26-35.	1.5	10
134	Arginine does not rescue p.Q188R mutation deleterious effect in classic galactosemia. Orphanet Journal of Rare Diseases, 2018, 13, 212.	2.7	9
135	Brain Phosphoinositide Extraction, Fractionation, and Analysis by MALDI-TOF MS. Methods in Molecular Biology, 2009, 579, 189-200.	0.9	9
136	Brain MRS glutamine as a biomarker to guide therapy of hyperammonemic coma. Molecular Genetics and Metabolism, 2017, 121, 9-15.	1.1	8
137	Transient developmental delays in infants with Duarte-2 variant galactosemia. Molecular Genetics and Metabolism, 2021, 134, 132-138.	1.1	8
138	A neurodevelopmental disorder caused by a novel de novo SVA insertion in exon 13 of the SRCAP gene. European Journal of Human Genetics, 2022, 30, 1083-1087.	2.8	8
139	Inborn Errors of Carbohydrate, Ammonia, Amino Acid, and Organic Acid Metabolism. , 2012, , 215-238.		7
140	Disorders of Galactose Metabolism. , 2015, , 615-626.		7
141	Hydroxysteroid 17-Beta Dehydrogenase Type 10 Disease in Siblings. JIMD Reports, 2016, 32, 25-32.	1.5	7
142	Conducting an investigator-initiated randomized double-blinded intervention trial in acute decompensation of inborn errors of metabolism: Lessons from the N-Carbamylglutamate Consortium. Translational Science of Rare Diseases, 2018, 3, 157-170.	1.5	7
143	Identification of neuronal structures and pathways corresponding to clinical functioning in galactosemia. Journal of Inherited Metabolic Disease, 2020, 43, 1205-1218.	3.6	7
144	Liquid Chromatographyâ€“Tandem Mass Spectrometry Enzyme Assay for UDP-Galactose 4â€²-Epimerase: Use of Fragment Intensity Ratio in Differentiation of Structural Isomers. Clinical Chemistry, 2014, 60, 783-790.	3.2	6

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145	Uridineâ€responsive epileptic encephalopathy due to inherited variants in CAD : A Tale of Two Siblings. Annals of Clinical and Translational Neurology, 2021, 8, 716-722.	3.7	6
146	A 9-Month-Old Boy with Seizures and Discrepant Urine Tryptophan Concentrations. Clinical Chemistry, 2011, 57, 545-548.	3.2	5
147	Acute Illness Protocol for Urea Cycle Disorders. Pediatric Emergency Care, 2018, 34, e115-e119.	0.9	5
148	Megaloblastic Anemia Progressing to Severe Thrombotic Microangiopathy in Patients with Disordered Vitamin B12 Metabolism: Case Reports and Literature Review. Journal of Pediatrics, 2018, 202, 315-319.e2.	1.8	5
149	Exome sequencing identifies novel missense and deletion variants in <scp>RTN4IP1</scp> associated with optic atrophy, global developmental delay, epilepsy, ataxia, and choreoathetosis. American Journal of Medical Genetics, Part A, 2021, 185, 203-207.	1.2	5
150	A 7-year old female with arthrogryposis multiplex congenita, Duane retraction syndrome, and Marcus Gunn phenomenon due to a ZC4H2 gene mutation: a clinical presentation of the Wieacker-Wolff syndrome. Ophthalmic Genetics, 2021, 42, 612-614.	1.2	5
151	Liver Failure as the Presentation of Ornithine Transcarbamylase Deficiency in a 13-Month-Old Female. JIMD Reports, 2017, 40, 17-22.	1.5	4
152	Biliary Atresia Associated With a Fatty Acid Oxidation Defect. Journal of Pediatric Gastroenterology and Nutrition, 2002, 35, 624-628.	1.8	3
153	Metabolic Profiling. Nestle Nutrition Workshop Series Paediatric Programme, 2008, 62, 55-80.	1.5	2
154	Back to the future: From genome to metabolome. Human Mutation, 2012, 33, 809-812.	2.5	2
155	Inborn Errors of Amino Acid and Organic Acid Metabolism. , 1998, , 799-819.		2
156	Results from a 78-week, single-arm, open-label Phase 2 study to evaluate UX007 in pediatric and adult patients with severe long-chain fatty acid oxidation disorders (LC-FAOD). Journal of Inherited Metabolic Disease, 2019, 42, 169.	3.6	2
157	Phenotypic variability in deficiency of the Î± subunit of succinate-CoA ligase. JIMD Reports, 2019, 46, 63-69.	1.5	2
158	The development of end stage renal disease in two patients with PMM2â€CDG. JIMD Reports, 2022, 63, 131-136.	1.5	2
159	Acidosis associated with dietotherapy of maple syrup urine disease. Journal of Pediatrics, 1980, 96, 62-64.	1.8	1
160	Concentration of White Blood Cell UDPGalactose and UDPGlucose Determined by High Performance Liquid Chromatography. Enzyme & Protein, 1993, 47, 105-115.	1.4	1
161	Inborn Errors of Carbohydrate, Ammonia, Amino Acid, and Organic Acid Metabolism. , 2005, , 227-257.		1
162	The unexplored potential of the pentose phosphate pathway in health and disease. Journal of Inherited Metabolic Disease, 2008, 31, 661-661.	3.6	1

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163	Commentary. Clinical Chemistry, 2010, 56, 1669-1669.	3.2	1
164	A 10-Month-Old With Intermittent Hypotonia and Paralysis. Pediatrics, 2016, 138, .	2.1	1
165	Phenotypic heterogeneity of a compound heterozygous SUCLA2 mutation. Molecular Genetics and Metabolism Reports, 2017, 12, 1.	1.1	1
166	Abnormal Glycerol Metabolism in a Child with Global Developmental Delay, Adrenal Insufficiency, and Intellectual Disability. Clinical Chemistry, 2018, 64, 1785-1787.	3.2	1
167	Hyperammonemia in a Child Presenting with Growth Delay, Short Stature, and Diarrhea. Clinical Chemistry, 2018, 64, 1260-1262.	3.2	1
168	Is Xâ€linked, infantile onset ALG13â€related developmental and epileptic encephalopathy a congenital disorder of glycosylation?. Epilepsia, 2021, 62, 335-336.	5.1	1
169	Clinical utility of brain MRS imaging of patients with adult-onset non-cirrhotic hyperammonemia. Molecular Genetics and Metabolism Reports, 2021, 27, 100742.	1.1	1
170	Introduction to the Metabolic and Biochemical Genetic Diseases. , 2005, , 217-226.		1
171	Inositol-Related Gene Knockouts Mimic Lithium's Effect on Mitochondrial Function. Nature Precedings, 2011, , .	0.1	0
172	Disorders of galactose metabolism. , 2020, , 803-816.		0
173	[13 C]â€galactose breath test in a patient with galactokinase deficiency and spastic diparesis. JIMD Reports, 2021, 59, 104-109.	1.5	0
174	Paroxysmal hyperthermia, dysautonomia and rhabdomyolysis in a patient with Leschâ€“Nyhan syndrome. JIMD Reports, 2021, 62, 30-34.	1.5	0
175	Pathophysiology of Metabolic Disease of the Liver. , 2004, , 1211-1218.		0
176	The neonatal brain mitochondrial ribosomal protein subunit 6 gene expression is normal in the lethal Smit1 knockout and independent of the rescue of the phenotype with prenatal myoâ€inositol treatment. FASEB Journal, 2008, 22, 805.13.	0.5	0
177	Inborn Errors of Amino Acid and Organic Acid Metabolism. , 1991, , 426-444.		0
178	Urea Cycle Disorders in the US and Europe â€“ Evidence-based Clinical Outcomes Derived from Two Decades of Experience with Prospective Registry Studies. , 2019, 50, .		0