Gerard Berry

List of Publications by Year
in descending order

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

#	Article	IF	Citations
1	Genetic Determinants of Sudden Unexpected Death in Pediatrics. Genetics in Medicine, 2022, 24, 839-850.	2.4	20
2	The development of end stage renal disease in two patients with PMM2â€CDG. JIMD Reports, 2022, 63, 131-136.	1.5	2
3	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
4	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
5	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
6	Galactokinase deficiency: lessons from the GalNet registry. Genetics in Medicine, 2021, 23, 202-210.	2.4	14
7	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
8	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
9	Is Xâ€linked, infantile onset ALG13â€related developmental and epileptic encephalopathy a congenital disorder of glycosylation?. Epilepsia, 2021, 62, 335-336.	5.1	1
10	[13 C]â€galactose breath test in a patient with galactokinase deficiency and spastic diparesis. JIMD Reports, 2021, 59, 104-109.	1.5	0
11	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
12	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
13	Transient developmental delays in infants with Duarte-2 variant galactosemia. Molecular Genetics and Metabolism, 2021, 134, 132-138.	1.1	8
14	Paroxysmal hyperthermia, dysautonomia and rhabdomyolysis in a patient with Leschâ€“Nyhan syndrome. JIMD Reports, 2021, 62, 30-34.	1.5	0
15	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
16	Defining a new immune deficiency syndrome: MAN2B2-CDG. Journal of Allergy and Clinical Immunology, 2020, 145, 1008-1011.	2.9	19
17	Disorders of galactose metabolism. , 2020, , 803-816.		0
18	The reâ€occurrence of cardiomyopathy in propionic acidemia after liver transplantation. JIMD Reports, 2020, 54, 3-8.	1.5	12

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19	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
20	A retrospective study of adult patients with noncirrhotic hyperammonemia. Journal of Inherited Metabolic Disease, 2020, 43, 1165-1172.	3.6	12
21	Identification of neuronal structures and pathways corresponding to clinical functioning in galactosemia. Journal of Inherited Metabolic Disease, 2020, 43, 1205-1218.	3.6	7
22	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
23	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
24	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
25	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
26	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
27	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
28	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
29	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
30	Phenotypic variability in deficiency of the Î± subunit of succinate-CoA ligase. JIMD Reports, 2019, 46, 63-69.	1.5	2
31	Hereditary galactosemia. Metabolism: Clinical and Experimental, 2018, 83, 188-196.	3.4	82
32	Impaired fertility and motor function in a zebrafish model for classic galactosemia. Journal of Inherited Metabolic Disease, 2018, 41, 117-127.	3.6	16
33	Inborn Errors of Metabolism with Hepatopathy. Pediatric Clinics of North America, 2018, 65, 337-352.	1.8	26
34	Urine oligosaccharide screening by MALDI-TOF for the identification of NGLY1 deficiency. Molecular Genetics and Metabolism, 2018, 124, 82-86.	1.1	29
35	Acute Illness Protocol for Urea Cycle Disorders. Pediatric Emergency Care, 2018, 34, e115-e119.	0.9	5
36	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

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37	Arginine does not rescue p.Q188R mutation deleterious effect in classic galactosemia. Orphanet Journal of Rare Diseases, 2018, 13, 212.	2.7	9
38	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
39	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
40	Abnormal Glycerol Metabolism in a Child with Global Developmental Delay, Adrenal Insufficiency, and Intellectual Disability. Clinical Chemistry, 2018, 64, 1785-1787.	3.2	1
41	KCTD7 deficiency defines a distinct neurodegenerative disorder with a conserved autophagyâ€lysosome defect. Annals of Neurology, 2018, 84, 766-780.	5.3	42
42	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
43	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
44	Hyperammonemia in a Child Presenting with Growth Delay, Short Stature, and Diarrhea. Clinical Chemistry, 2018, 64, 1260-1262.	3.2	1
45	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
46	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
47	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
48	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
49	Phenotypic heterogeneity of a compound heterozygous SUCLA2 mutation. Molecular Genetics and Metabolism Reports, 2017, 12, 1.	1.1	1
50	Brain MRS glutamine as a biomarker to guide therapy of hyperammonemic coma. Molecular Genetics and Metabolism, 2017, 121, 9-15.	1.1	8
51	Liver Failure as the Presentation of Ornithine Transcarbamylase Deficiency in a 13-Month-Old Female. JIMD Reports, 2017, 40, 17-22.	1.5	4
52	Acute Illness Protocol for Organic Acidemias. Pediatric Emergency Care, 2017, 33, 142-146.	0.9	17
53	De Novo TUBB2A Variant Presenting With Anterior Temporal Pachygyria. Journal of Child Neurology, 2017, 32, 127-131.	1.4	15
54	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

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55	Fertility in adult women with classic galactosemia and primary ovarian insufficiency. Fertility and Sterility, 2017, 108, 168-174.	1.0	42
56	Hydroxysteroid 17-Beta Dehydrogenase Type 10 Disease in Siblings. JIMD Reports, 2016, 32, 25-32.	1.5	7
57	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	2.5	40
58	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
59	A novel de novo mutation inATP1A3and childhood-onset schizophrenia. Journal of Physical Education and Sports Management, 2016, 2, a001008.	1.2	46
60	A 10-Month-Old With Intermittent Hypotonia and Paralysis. Pediatrics, 2016, 138, .	2.1	1
61	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
62	Disease Heterogeneity in Na+/Citrate Cotransporter Deficiency. JIMD Reports, 2016, 31, 107-111.	1.5	11
63	Galactose metabolism and health. Current Opinion in Clinical Nutrition and Metabolic Care, 2015, 18, 422-427.	2.5	162
64	Disorders of Galactose Metabolism. , 2015, , 615-626.		7
65	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
66	Inositol-Related Gene Knockouts Mimic Lithiumâ€™s Effect on Mitochondrial Function. Neuropsychopharmacology, 2014, 39, 319-328.	5.4	42
67	KCNQ1, KCNE2, and Na ⁺ -Coupled Solute Transporters Form Reciprocally Regulating Complexes That Affect Neuronal Excitability. Science Signaling, 2014, 7, ra22.	3.6	56
68	A compound heterozygous mutation in GPD1 causes hepatomegaly, steatohepatitis, and hypertriglyceridemia. European Journal of Human Genetics, 2014, 22, 1229-1232.	2.8	38
69	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
70	Copy number variation plays an important role in clinical epilepsy. Annals of Neurology, 2014, 75, 943-958.	5.3	147
71	Methylmalonic acidemia: A megamitochondrial disorder affecting the kidney. Pediatric Nephrology, 2014, 29, 2139-2146.	1.7	52
72	Uremic solutes and risk of end-stage renal disease in type 2 diabetes: metabolomic study. Kidney International, 2014, 85, 1214-1224.	5.2	182

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73	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
74	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
75	Fertility preservation in female classic galactosemia patients. Orphanet Journal of Rare Diseases, 2013, 8, 107.	2.7	34
76	The male reproductive system in classic galactosemia: cryptorchidism and low semen volume. Journal of Inherited Metabolic Disease, 2013, 36, 779-786.	3.6	24
77	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
78	Targeted exome sequencing of suspected mitochondrial disorders. Neurology, 2013, 80, 1762-1770.	1.1	155
79	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
80	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
81	Bumetanide Enhances Phenobarbital Efficacy in a Rat Model of Hypoxic Neonatal Seizures. PLoS ONE, 2013, 8, e57148.	2.5	117
82	Homozygous PLCB1 deletion associated with malignant migrating partial seizures in infancy. Epilepsia, 2012, 53, e146-50.	5.1	104
83	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
84	Recurrent unexplained hyperammonemia in an adolescent with arginase deficiency. Clinical Biochemistry, 2012, 45, 1583-1586.	1.9	10
85	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
86	N- and O-linked glycosylation of total plasma glycoproteins in galactosemia. Molecular Genetics and Metabolism, 2012, 106, 442-454.	1.1	34
87	Exploring concordance and discordance for return of incidental findings from clinical sequencing. Genetics in Medicine, 2012, 14, 405-410.	2.4	149
88	Galactosemia: When is it a newborn screening emergency?. Molecular Genetics and Metabolism, 2012, 106, 7-11.	1.1	68
89	Disorders of Galactose Metabolism. , 2012, , 141-150.		14
90	Inborn Errors of Carbohydrate, Ammonia, Amino Acid, and Organic Acid Metabolism. , 2012, , 215-238.		7

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91	Back to the future: From genome to metabolome. Human Mutation, 2012, 33, 809-812.	2.5	2
92	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
93	The adult galactosemic phenotype. Journal of Inherited Metabolic Disease, 2012, 35, 279-286.	3.6	151
94	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
95	Inositol-Related Gene Knockouts Mimic Lithium's Effect on Mitochondrial Function. Nature Precedings, 2011, , .	0.1	0
96	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
97	Is prenatal myoâ€inositol deficiency a mechanism of CNS injury in galactosemia?. Journal of Inherited Metabolic Disease, 2011, 34, 345-355.	3.6	46
98	Psychosocial developmental milestones in men with classic galactosemia. Journal of Inherited Metabolic Disease, 2011, 34, 415-419.	3.6	22
99	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
100	A 9-Month-Old Boy with Seizures and Discrepant Urine Tryptophan Concentrations. Clinical Chemistry, 2011, 57, 545-548.	3.2	5
101	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
102	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
103	Quantification of Galactose-1-Phosphate Uridyltransferase Enzyme Activity by Liquid Chromatographyâ€“Tandem Mass Spectrometry. Clinical Chemistry, 2010, 56, 772-780.	3.2	22
104	Commentary. Clinical Chemistry, 2010, 56, 1669-1669.	3.2	1
105	Knockout mice in understanding the mechanism of action of lithium. Biochemical Society Transactions, 2009, 37, 1121-1125.	3.4	48
106	Brain Phosphoinositide Extraction, Fractionation, and Analysis by MALDI-TOF MS. Methods in Molecular Biology, 2009, 579, 189-200.	0.9	9
107	The unexplored potential of the pentose phosphate pathway in health and disease. Journal of Inherited Metabolic Disease, 2008, 31, 661-661.	3.6	1
108	Galactosemia and Amenorrhea in the Adolescent. Annals of the New York Academy of Sciences, 2008, 1135, 112-117.	3.8	34

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109	Homozygote inositol transporter knockout mice show a lithiumâ€like phenotype. Bipolar Disorders, 2008, 10, 453-459.	1.9	29
110	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
111	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
112	Metabolic Profiling. Nestle Nutrition Workshop Series Paediatric Programme, 2008, 62, 55-80.	1.5	2
113	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
114	Survival after Treatment with Phenylacetate and Benzoate for Urea-Cycle Disorders. New England Journal of Medicine, 2007, 356, 2282-2292.	27.0	320
115	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
116	Epimerase-Deficiency Galactosemia Is Not a Binary Condition. American Journal of Human Genetics, 2006, 78, 89-102.	6.2	77
117	SMIT1 haploinsufficiency causes brain inositol deficiency without affecting lithium-sensitive behavior. Molecular Genetics and Metabolism, 2006, 88, 384-388.	1.1	32
118	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
119	Disorders of Galactose Metabolism. , 2006, , 121-130.		69
120	Inborn Errors of Carbohydrate, Ammonia, Amino Acid, and Organic Acid Metabolism. , 2005, , 227-257.		1
121	Elements of diabetic nephropathy in a patient with GLUT2 deficiency. Molecular Genetics and Metabolism, 2005, 86, 473-477.	1.1	17
122	Introduction to the Metabolic and Biochemical Genetic Diseases. , 2005, , 217-226.		1
123	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
124	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
125	Extended [13C]galactose oxidation studies in patients with galactosemia. Molecular Genetics and Metabolism, 2004, 82, 130-136.	1.1	20
126	Pathophysiology of Metabolic Disease of the Liver. , 2004, , 1211-1218.		0

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127	MR diffusion imaging and MR spectroscopy of maple syrup urine disease during acute metabolic decompensation. Neuroradiology, 2003, 45, 393-399.	2.2	157
128	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
129	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
130	In vivo pyruvate detected by MR spectroscopy in neonatal pyruvate dehydrogenase deficiency. American Journal of Neuroradiology, 2003, 24, 1471-4.	2.4	39
131	Biliary Atresia Associated With a Fatty Acid Oxidation Defect. Journal of Pediatric Gastroenterology and Nutrition, 2002, 35, 624-628.	1.8	3
132	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
133	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
134	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
135	Ornithine transcarbamylase deficiency and pancreatitis. Journal of Pediatrics, 2001, 138, 123-124.	1.8	12
136	In vivo evidence of brain galactitol accumulation in an infant with galactosemia and encephalopathy. Journal of Pediatrics, 2001, 138, 260-262.	1.8	68
137	Long-term management of patients with urea cycle disorders. Journal of Pediatrics, 2001, 138, S56-S61.	1.8	62
138	Proton magnetic resonance spectroscopy of brain metabolites in galactosemia. Annals of Neurology, 2001, 50, 266-269.	5.3	40
139	Galactose Breath Testing Distinguishes Variant and Severe Galactose-1-Phosphate Uridyltransferase Genotypes. Pediatric Research, 2000, 48, 323-328.	2.3	44
140	Galactose Metabolism by the Mouse with Galactose-1-Phosphate Uridyltransferase Deficiency. Pediatric Research, 2000, 48, 211-217.	2.3	54
141	Fatal Hyperammonemia after Orthotopic Lung Transplantation. Annals of Internal Medicine, 2000, 132, 283.	3.9	76
142	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
143	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
144	Risk factors for premature ovarian failure in females with galactosemia. Journal of Pediatrics, 2000, 137, 833-841.	1.8	99

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145	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
146	Urine and plasma galactitol in patients with galactose-1-phosphate uridyltransferase deficiency galactosemia. Metabolism: Clinical and Experimental, 1999, 48, 1294-1302.	3.4	45
147	In vivo brain myo -inositol levels in children with Down syndrome. Journal of Pediatrics, 1999, 135, 94-97.	1.8	39
148	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
149	Inborn Errors of Amino Acid and Organic Acid Metabolism. , 1998, , 799-819.		2
150	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
151	A syndrome of congenital hyperinsulinism and hyperammonemia. Journal of Pediatrics, 1997, 130, 661-664.	1.8	101
152	Urinary Galactonate in Patients with Galactosemia: Quantitation by Nuclear Magnetic Resonance Spectroscopy. Pediatric Research, 1997, 42, 855-861.	2.3	52
153	Diabetes-like renal glomerular disease in Fanconi-Bickel syndrome. Pediatric Nephrology, 1995, 9, 287-291.	1.7	43
154	The role of polyols in the pathophysiology of hypergalactosemia. European Journal of Pediatrics, 1995, 154, S53-S64.	2.7	61
155	The human osmoregulatory Na+/myo-inositol cotransporter gene (SLC5A3): molecular cloning and localization to chromosome 21. Genomics, 1995, 25, 507-513.	2.9	119
156	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
157	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
158	Kinetic evidence for compartmentalization of myo-inositol in hepatocytes. Metabolism: Clinical and Experimental, 1993, 42, 395-401.	3.4	19
159	Renal Handling of Carnitine in Secondary Carnitine Deficiency Disorders. Pediatric Research, 1993, 34, 89-96.	2.3	40
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	A Deficiency of Carnitineâ€“Acylcarnitine Translocase in the Inner Mitochondrial Membrane. New England Journal of Medicine, 1992, 327, 19-23.	27.0	183
162	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

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163	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
164	Intellectual outcome in children with maple syrup urine disease. Journal of Pediatrics, 1991, 119, 46-50.	1.8	81
165	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
166	Cytochrome c oxidase-associated Leigh syndrome: Phenotypic features and pathogenetic speculations. Journal of the Neurological Sciences, 1991, 104, 97-111.	0.6	108
167	The concentration of red blood cell UDPGlucose and UDPGalactose determined by high-performance liquid chromatography. Analytical Biochemistry, 1991, 194, 388-393.	2.4	42
168	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
169	Inborn Errors of Amino Acid and Organic Acid Metabolism. , 1991, , 426-444.		0
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