

Gerard Berry

List of Publications by Year in descending order

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178
papers

7,597
citations

46984

47
h-index

66879

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. <i>New England Journal of Medicine</i> , 2007, 356, 2282-2292.	13.9	320
2	Deletions of <i>NRXN1</i> (<i>neurexin1</i>) predispose to a wide spectrum of developmental disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 937-947.	1.1	217
3	A Deficiency of Carnitine- <i>Acylcarnitine</i> Translocase in the Inner Mitochondrial Membrane. <i>New England Journal of Medicine</i> , 1992, 327, 19-23.	13.9	183
4	Uremic solutes and risk of end-stage renal disease in type 2 diabetes: metabolomic study. <i>Kidney International</i> , 2014, 85, 1214-1224.	2.6	182
5	Galactose metabolism and health. <i>Current Opinion in Clinical Nutrition and Metabolic Care</i> , 2015, 18, 422-427.	1.3	162
6	MR diffusion imaging and MR spectroscopy of maple syrup urine disease during acute metabolic decompensation. <i>Neuroradiology</i> , 2003, 45, 393-399.	1.1	157
7	Transcription of the Sodium/myo-Inositol Cotransporter Gene Is Regulated by Multiple Tonicity-responsive Enhancers Spread over 50 Kilobase Pairs in the 5'-Flanking Region. <i>Journal of Biological Chemistry</i> , 1998, 273, 20615-20621.	1.6	155
8	Targeted exome sequencing of suspected mitochondrial disorders. <i>Neurology</i> , 2013, 80, 1762-1770.	1.5	155
9	The adult galactosemic phenotype. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 279-286.	1.7	151
10	Exploring concordance and discordance for return of incidental findings from clinical sequencing. <i>Genetics in Medicine</i> , 2012, 14, 405-410.	1.1	149
11	The mutation spectrum of the facilitative glucose transporter gene <i>SLC2A2</i> (<i>GLUT2</i>) in patients with Fanconi-Bickel syndrome. <i>Human Genetics</i> , 2002, 110, 21-29.	1.8	147
12	Copy number variation plays an important role in clinical epilepsy. <i>Annals of Neurology</i> , 2014, 75, 943-958.	2.8	147
13	Acute extrapyramidal syndrome in methylmalonic acidemia: "Metabolic stroke" involving the globus pallidus. <i>Journal of Pediatrics</i> , 1988, 113, 1022-1027.	0.9	135
14	International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 171-176.	1.7	132
15	The human osmoregulatory Na ⁺ /myo-inositol cotransporter gene (<i>SLC5A3</i>): molecular cloning and localization to chromosome 21. <i>Genomics</i> , 1995, 25, 507-513.	1.3	119
16	Bumetanide Enhances Phenobarbital Efficacy in a Rat Model of Hypoxic Neonatal Seizures. <i>PLoS ONE</i> , 2013, 8, e57148.	1.1	117
17	Cytochrome c oxidase-associated Leigh syndrome: Phenotypic features and pathogenetic speculations. <i>Journal of the Neurological Sciences</i> , 1991, 104, 97-111.	0.3	108
18	Homozygous <i>PLCB1</i> deletion associated with malignant migrating partial seizures in infancy. <i>Epilepsia</i> , 2012, 53, e146-50.	2.6	104

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19	A syndrome of congenital hyperinsulinism and hyperammonemia. <i>Journal of Pediatrics</i> , 1997, 130, 661-664.	0.9	101
20	Risk factors for premature ovarian failure in females with galactosemia. <i>Journal of Pediatrics</i> , 2000, 137, 833-841.	0.9	99
21	Targeting proximal tubule mitochondrial dysfunction attenuates the renal disease of methylmalonic acidemia. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 13552-13557.	3.3	97
22	Glycogen synthase kinase-3 is essential for β -arrestin-2 complex formation and lithium-sensitive behaviors in mice. <i>Journal of Clinical Investigation</i> , 2011, 121, 3756-3762.	3.9	96
23	Newborn Screening by Tandem Mass Spectrometry for Medium-Chain Acyl-CoA Dehydrogenase Deficiency: A Cost-Effectiveness Analysis. <i>Pediatrics</i> , 2003, 112, 1005-1015.	1.0	90
24	High-performance liquid chromatography of phospholipids with UV detection: optimization of separations on silica. <i>Biomedical Applications</i> , 1981, 225, 319-328.	1.7	86
25	Hereditary galactosemia. <i>Metabolism: Clinical and Experimental</i> , 2018, 83, 188-196.	1.5	82
26	Intellectual outcome in children with maple syrup urine disease. <i>Journal of Pediatrics</i> , 1991, 119, 46-50.	0.9	81
27	Loss of Murine Na ⁺ /myo-Inositol Cotransporter Leads to Brain myo-Inositol Depletion and Central Apnea. <i>Journal of Biological Chemistry</i> , 2003, 278, 18297-18302.	1.6	80
28	Epimerase-Deficiency Galactosemia Is Not a Binary Condition. <i>American Journal of Human Genetics</i> , 2006, 78, 89-102.	2.6	77
29	Diversity of approaches to classic galactosemia around the world: a comparison of diagnosis, intervention, and outcomes. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 1037-1049.	1.7	77
30	Fatal Hyperammonemia after Orthotopic Lung Transplantation. <i>Annals of Internal Medicine</i> , 2000, 132, 283.	2.0	76
31	The rate of de novo galactose synthesis in patients with galactose-1-phosphate uridylyltransferase deficiency. <i>Molecular Genetics and Metabolism</i> , 2004, 81, 22-30.	0.5	75
32	Liver transplantation is not curative for methylmalonic acidopathy caused by methylmalonyl-CoA mutase deficiency. <i>Molecular Genetics and Metabolism</i> , 2006, 88, 322-326.	0.5	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. <i>Journal of Pediatrics</i> , 2001, 138, 260-262.	0.9	68
35	Galactosemia: When is it a newborn screening emergency?. <i>Molecular Genetics and Metabolism</i> , 2012, 106, 7-11.	0.5	68
36	Sodium-myoinositol cotransporter-1, SMIT1, mediates the production of reactive oxygen species induced by hyperglycemia in the heart. <i>Scientific Reports</i> , 2017, 7, 41166.	1.6	64

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37	Long-term management of patients with urea cycle disorders. <i>Journal of Pediatrics</i> , 2001, 138, S56-S61.	0.9	62
38	The role of polyols in the pathophysiology of hypergalactosemia. <i>European Journal of Pediatrics</i> , 1995, 154, S53-S64.	1.3	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. <i>Molecular Genetics and Metabolism</i> , 2008, 93, 179-189.	0.5	61
40	DDOST Mutations Identified by Whole-Exome Sequencing Are Implicated in Congenital Disorders of Glycosylation. <i>American Journal of Human Genetics</i> , 2012, 90, 363-368.	2.6	60
41	Branched-Chain Amino Acid-Free Parenteral Nutrition in the Treatment of Acute Metabolic Decompensation in Patients with Maple Syrup Urine Disease. <i>New England Journal of Medicine</i> , 1991, 324, 175-179.	13.9	56
42	KCNQ1, KCNE2, and Na ⁺ -Coupled Solute Transporters Form Reciprocally Regulating Complexes That Affect Neuronal Excitability. <i>Science Signaling</i> , 2014, 7, ra22.	1.6	56
43	Galactose Metabolism by the Mouse with Galactose-1-Phosphate Uridyltransferase Deficiency. <i>Pediatric Research</i> , 2000, 48, 211-217.	1.1	54
44	Urinary Galactonate in Patients with Galactosemia: Quantitation by Nuclear Magnetic Resonance Spectroscopy. <i>Pediatric Research</i> , 1997, 42, 855-861.	1.1	52
45	Methylmalonic acidemia: A megamitochondrial disorder affecting the kidney. <i>Pediatric Nephrology</i> , 2014, 29, 2139-2146.	0.9	52
46	Phosphoinositide deficiency due to inositol depletion is not a mechanism of lithium action in brain. <i>Molecular Genetics and Metabolism</i> , 2004, 82, 87-92.	0.5	51
47	Whole genome sequencing identifies <i>SCN2A</i> mutation in monozygotic twins with Ohtahara syndrome and unique neuropathologic findings. <i>Epilepsia</i> , 2013, 54, e81-5.	2.6	49
48	A re-evaluation of life-long severe galactose restriction for the nutrition management of classic galactosemia. <i>Molecular Genetics and Metabolism</i> , 2014, 112, 191-197.	0.5	49
49	Knockout mice in understanding the mechanism of action of lithium. <i>Biochemical Society Transactions</i> , 2009, 37, 1121-1125.	1.6	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. <i>Genomics</i> , 2000, 68, 144-151.	1.3	47
51	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 (LCFAOD). <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 169-177.	1.7	47
52	Is prenatal myo-inositol deficiency a mechanism of CNS injury in galactosemia?. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 345-355.	1.7	46
53	A novel de novo mutation in <i>ATP1A3</i> and childhood-onset schizophrenia. <i>Journal of Physical Education and Sports Management</i> , 2016, 2, a001008.	0.5	46
54	Isovaleric acidemia: Medical and neurodevelopmental effects of long-term therapy. <i>Journal of Pediatrics</i> , 1988, 113, 58-64.	0.9	45

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55	Urine and plasma galactitol in patients with galactose-1-phosphate uridyltransferase deficiency galactosemia. <i>Metabolism: Clinical and Experimental</i> , 1999, 48, 1294-1302.	1.5	45
56	Galactose Breath Testing Distinguishes Variant and Severe Galactose-1-Phosphate Uridyltransferase Genotypes. <i>Pediatric Research</i> , 2000, 48, 323-328.	1.1	44
57	Sensitive isotope dilution liquid chromatography/tandem mass spectrometry method for quantitative analysis of bumetanide in serum and brain tissue. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2011, 879, 998-1002.	1.2	44
58	Diabetes-like renal glomerular disease in Fanconi-Bickel syndrome. <i>Pediatric Nephrology</i> , 1995, 9, 287-291.	0.9	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. <i>Molecular Genetics and Metabolism</i> , 2008, 95, 81-95.	0.5	43
60	The concentration of red blood cell UDPGlucose and UDPGalactose determined by high-performance liquid chromatography. <i>Analytical Biochemistry</i> , 1991, 194, 388-393.	1.1	42
61	Inositol-Related Gene Knockouts Mimic Lithium's Effect on Mitochondrial Function. <i>Neuropsychopharmacology</i> , 2014, 39, 319-328.	2.8	42
62	<i>KCTD7</i> deficiency defines a distinct neurodegenerative disorder with a conserved autophagy-lysosome defect. <i>Annals of Neurology</i> , 2018, 84, 766-780.	2.8	42
63	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. <i>Annals of Neurology</i> , 2019, 86, 116-128.	2.8	42
64	Fertility in adult women with classic galactosemia and primary ovarian insufficiency. <i>Fertility and Sterility</i> , 2017, 108, 168-174.	0.5	42
65	Renal Handling of Carnitine in Secondary Carnitine Deficiency Disorders. <i>Pediatric Research</i> , 1993, 34, 89-96.	1.1	40
66	Proton magnetic resonance spectroscopy of brain metabolites in galactosemia. <i>Annals of Neurology</i> , 2001, 50, 266-269.	2.8	40
67	Introduction to the Maastricht workshop: lessons from the past and new directions in galactosemia. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 249-255.	1.7	40
68	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. <i>Human Mutation</i> , 2016, 37, 653-660.	1.1	40
69	The Structural Organization of the Human Na ⁺ /Myo-inositol Cotransporter (SLC5A3) Gene and Characterization of the Promoter. <i>Genomics</i> , 1997, 46, 459-465.	1.3	39
70	In vivo brain myo -inositol levels in children with Down syndrome. <i>Journal of Pediatrics</i> , 1999, 135, 94-97.	0.9	39
71	In vivo pyruvate detected by MR spectroscopy in neonatal pyruvate dehydrogenase deficiency. <i>American Journal of Neuroradiology</i> , 2003, 24, 1471-4.	1.2	39
72	Evidence for Alternate Galactose Oxidation in a Patient with Deletion of the Galactose-1-Phosphate Uridyltransferase Gene. <i>Molecular Genetics and Metabolism</i> , 2001, 72, 316-321.	0.5	38

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73	A compound heterozygous mutation in GPD1 causes hepatomegaly, steatohepatitis, and hypertriglyceridemia. <i>European Journal of Human Genetics</i> , 2014, 22, 1229-1232.	1.4	38
74	Novel founder intronic variant in SLC39A14 in two families causing Manganism and potential treatment strategies. <i>Molecular Genetics and Metabolism</i> , 2018, 124, 161-167.	0.5	36
75	Effects of triheptanoin (<sc>UX007</sc>) in patients with longâ€chain fatty acid oxidation disorders: Results from an <sc>openâ€label</sc>, <sc>longâ€term</sc> extension study. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 253-263.	1.7	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. <i>Analytical Biochemistry</i> , 2007, 362, 155-167.	1.1	34
77	<i>Galactosemia and Amenorrhea in the Adolescent</i>. <i>Annals of the New York Academy of Sciences</i> , 2008, 1135, 112-117.	1.8	34
78	N- and O-linked glycosylation of total plasma glycoproteins in galactosemia. <i>Molecular Genetics and Metabolism</i> , 2012, 106, 442-454.	0.5	34
79	Fertility preservation in female classic galactosemia patients. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 107.	1.2	34
80	Red blood cell uridine sugar nucleotide levels in patients with classic galactosemia and other metabolic disorders. <i>Metabolism: Clinical and Experimental</i> , 1992, 41, 783-787.	1.5	32
81	SMIT1 haploinsufficiency causes brain inositol deficiency without affecting lithium-sensitive behavior. <i>Molecular Genetics and Metabolism</i> , 2006, 88, 384-388.	0.5	32
82	Galactose Metabolism in Mice with Galactose-1-Phosphate Uridyltransferase Deficiency: Sucklings and 7-Week-Old Animals Fed a High-Galactose Diet. <i>Molecular Genetics and Metabolism</i> , 2001, 72, 306-315.	0.5	30
83	Homozygote inositol transporter knockout mice show a lithiumâ€like phenotype. <i>Bipolar Disorders</i> , 2008, 10, 453-459.	1.1	29
84	Urine oligosaccharide screening by MALDI-TOF for the identification of NGLY1 deficiency. <i>Molecular Genetics and Metabolism</i> , 2018, 124, 82-86.	0.5	29
85	Galactose-1 phosphate uridylyltransferase (GalT) gene: A novel positive regulator of the PI3K/Akt signaling pathway in mouse fibroblasts. <i>Biochemical and Biophysical Research Communications</i> , 2016, 470, 205-212.	1.0	28
86	Phenotypic expansion of <sc>Boschâ€Boonstraâ€Schaaf</sc> optic atrophy syndrome and further evidence for genotypeâ€phenotype correlations. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1426-1437.	0.7	27
87	³¹ P NMR analysis of red blood cell UDPGlucose and UDPGalactose: Comparison with HPLC and enzymatic methods. <i>Analytical Biochemistry</i> , 1992, 202, 105-110.	1.1	26
88	Inborn Errors of Metabolism with Hepatopathy. <i>Pediatric Clinics of North America</i> , 2018, 65, 337-352.	0.9	26
89	Gainâ€ofâ€function variants in the <i>ODC1</i> gene cause a syndromic neurodevelopmental disorder associated with macrocephaly, alopecia, dysmorphic features, and neuroimaging abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2554-2560.	0.7	26
90	Pathophysiology and targets for treatment in hereditary galactosemia: A systematic review of animal and cellular models. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 392-408.	1.7	25

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91	The male reproductive system in classic galactosemia: cryptorchidism and low semen volume. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 779-786.	1.7	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. <i>Molecular Genetics and Metabolism</i> , 2017, 120, 213-222.	0.5	24
93	High-performance liquid chromatography of phospholipids: Quantitation by phosphate analysis. <i>Analytical Biochemistry</i> , 1983, 135, 239-243.	1.1	23
94	Quantification of Galactose-1-Phosphate Uridyltransferase Enzyme Activity by Liquid Chromatography-Tandem Mass Spectrometry. <i>Clinical Chemistry</i> , 2010, 56, 772-780.	1.5	22
95	Psychosocial developmental milestones in men with classic galactosemia. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 415-419.	1.7	22
96	Use of citrulline as a diagnostic marker in the prospective treatment of urea cycle disorders. <i>Journal of Pediatrics</i> , 1991, 118, 914-917.	0.9	21
97	Comparison of erythrocyte uridine sugar nucleotide levels in normals, classic galactosemics, and patients with other metabolic disorders. <i>Metabolism: Clinical and Experimental</i> , 1995, 44, 597-604.	1.5	21
98	Galactose Content of Legumes, Caseinates, and Some Hard Cheeses: Implications for Diet Treatment of Classic Galactosemia. <i>Journal of Agricultural and Food Chemistry</i> , 2014, 62, 1397-1402.	2.4	21
99	Extended [13C]galactose oxidation studies in patients with galactosemia. <i>Molecular Genetics and Metabolism</i> , 2004, 82, 130-136.	0.5	20
100	Ultra fast and sensitive liquid chromatography tandem mass spectrometry based assay for galactose-1-phosphate uridylyltransferase and galactokinase deficiencies. <i>Molecular Genetics and Metabolism</i> , 2011, 102, 33-40.	0.5	20
101	Genetic Determinants of Sudden Unexpected Death in Pediatrics. <i>Genetics in Medicine</i> , 2022, 24, 839-850.	1.1	20
102	Kinetic evidence for compartmentalization of myo-inositol in hepatocytes. <i>Metabolism: Clinical and Experimental</i> , 1993, 42, 395-401.	1.5	19
103	Novel variants in <i>SPTAN1</i> without epilepsy: An expansion of the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2768-2776.	0.7	19
104	Defining a new immune deficiency syndrome: MAN2B2-CDG. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 145, 1008-1011.	1.5	19
105	CMP-dependent phosphatidylinositol:myo-inositol exchange activity in isolated nerve-endings. <i>Biochemical and Biophysical Research Communications</i> , 1983, 112, 817-821.	1.0	18
106	Rhabdomyolysis, acute renal failure, and cardiac arrest secondary to status dystonicus in a child with glutaric aciduria type I. <i>Molecular Genetics and Metabolism</i> , 2012, 106, 488-490.	0.5	18
107	5,10-methenyltetrahydrofolate synthetase deficiency causes a neurometabolic disorder associated with microcephaly, epilepsy, and cerebral hypomyelination. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 118-126.	0.5	18
108	Elements of diabetic nephropathy in a patient with GLUT2 deficiency. <i>Molecular Genetics and Metabolism</i> , 2005, 86, 473-477.	0.5	17

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109	Witnessed sleep-related seizure and sudden unexpected death in infancy: a case report. <i>Forensic Science, Medicine, and Pathology</i> , 2013, 9, 418-421.	0.6	17
110	Management of a Woman With Maple Syrup Urine Disease During Pregnancy, Delivery, and Lactation. <i>Journal of Parenteral and Enteral Nutrition</i> , 2015, 39, 875-879.	1.3	17
111	Acute Illness Protocol for Organic Acidemias. <i>Pediatric Emergency Care</i> , 2017, 33, 142-146.	0.5	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. <i>Journal of Neurochemistry</i> , 1986, 46, 1073-1080.	2.1	16
113	N-Acetylcysteine Therapy in an Infant with Transaldolase Deficiency Is Well Tolerated and Associated with Normalization of Alpha Fetoprotein Levels. <i>JIMD Reports</i> , 2016, 31, 73-77.	0.7	16
114	Impaired fertility and motor function in a zebrafish model for classic galactosemia. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 117-127.	1.7	16
115	Apparent Galactose Appearance Rate in Human Galactosemia Based on Plasma [13C]Galactose Isotopic Enrichment. <i>Molecular Genetics and Metabolism</i> , 2000, 70, 261-271.	0.5	15
116	De Novo <i>TUBB2A</i> Variant Presenting With Anterior Temporal Pachygyria. <i>Journal of Child Neurology</i> , 2017, 32, 127-131.	0.7	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. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 147-154.	0.5	14
119	An emerging role for endothelial barrier support therapy for congenital disorders of glycosylation. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 880-890.	1.7	14
120	Galactokinase deficiency: lessons from the GalNet registry. <i>Genetics in Medicine</i> , 2021, 23, 202-210.	1.1	14
121	Abnormal myo-inositol and phospholipid metabolism in cultured fibroblasts from patients with ataxia telangiectasia. <i>Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids</i> , 1999, 1437, 287-300.	1.2	12
122	Ornithine transcarbamylase deficiency and pancreatitis. <i>Journal of Pediatrics</i> , 2001, 138, 123-124.	0.9	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. <i>Molecular Genetics and Metabolism Reports</i> , 2018, 16, 23-29.	0.4	12
124	The re-occurrence of cardiomyopathy in propionic acidemia after liver transplantation. <i>JIMD Reports</i> , 2020, 54, 3-8.	0.7	12
125	A retrospective study of adult patients with noncirrhotic hyperammonemia. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1165-1172.	1.7	12
126	Pathophysiology of long-term complications in classic galactosemia: What we do and do not know. <i>Molecular Genetics and Metabolism</i> , 2022, 137, 33-39.	0.5	12

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127	Uridine Diphosphate Hexoses in Leukocytes and Fibroblasts of Classic Galactosemics and Patients with Other Metabolic Diseases. <i>Pediatric Research</i> , 1994, 36, 613-618.	1.1	11
128	Disease Heterogeneity in Na ⁺ /Citrate Cotransporter Deficiency. <i>JIMD Reports</i> , 2016, 31, 107-111.	0.7	11
129	The ability of an LC-MS/MS-based erythrocyte GALT enzyme assay to predict the phenotype in subjects with GALT deficiency. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 368-376.	0.5	11
130	Experimental Galactose Toxicity: Effects on Synaptosomal Phosphatidylinositol Metabolism. <i>Journal of Neurochemistry</i> , 1981, 37, 888-891.	2.1	10
131	Uridine diphosphate glucose and uridine diphosphate galactose in galactosemia. <i>Journal of Pediatrics</i> , 1990, 117, 838-840.	0.9	10
132	Recurrent unexplained hyperammonemia in an adolescent with arginase deficiency. <i>Clinical Biochemistry</i> , 2012, 45, 1583-1586.	0.8	10
133	A novel null mutation in the pyruvate dehydrogenase phosphatase catalytic subunit gene (PDP1) causing pyruvate dehydrogenase complex deficiency. <i>JIMD Reports</i> , 2019, 48, 26-35.	0.7	10
134	Arginine does not rescue p.Q188R mutation deleterious effect in classic galactosemia. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 212.	1.2	9
135	Brain Phosphoinositide Extraction, Fractionation, and Analysis by MALDI-TOF MS. <i>Methods in Molecular Biology</i> , 2009, 579, 189-200.	0.4	9
136	Brain MRS glutamine as a biomarker to guide therapy of hyperammonemic coma. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 9-15.	0.5	8
137	Transient developmental delays in infants with Duarte-2 variant galactosemia. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 132-138.	0.5	8
138	A neurodevelopmental disorder caused by a novel de novo SVA insertion in exon 13 of the SRCAP gene. <i>European Journal of Human Genetics</i> , 2022, 30, 1083-1087.	1.4	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. <i>JIMD Reports</i> , 2016, 32, 25-32.	0.7	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. <i>Translational Science of Rare Diseases</i> , 2018, 3, 157-170.	1.6	7
143	Identification of neuronal structures and pathways corresponding to clinical functioning in galactosemia. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1205-1218.	1.7	7
144	Liquid Chromatography-Tandem Mass Spectrometry Enzyme Assay for UDP-Galactose 4-epimerase: Use of Fragment Intensity Ratio in Differentiation of Structural Isomers. <i>Clinical Chemistry</i> , 2014, 60, 783-790.	1.5	6

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145	Uridine-responsive epileptic encephalopathy due to inherited variants in CAD : A Tale of Two Siblings. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 716-722.	1.7	6
146	A 9-Month-Old Boy with Seizures and Discrepant Urine Tryptophan Concentrations. <i>Clinical Chemistry</i> , 2011, 57, 545-548.	1.5	5
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