

Peter Theodore Clayton

List of Publications by Year in descending order

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142
papers

10,009
citations

30070

54
h-index

39675

94
g-index

150
all docs

150
docs citations

150
times ranked

10061
citing authors

#	ARTICLE	IF	CITATIONS
1	Tissue Proteome of 2-Hydroxyacyl-CoA Lyase Deficient Mice Reveals Peroxisome Proliferation and Activation of Î±-Oxidation. International Journal of Molecular Sciences, 2022, 23, 987.	4.1	4
2	Bile acid biosynthesis in Smith-Lemli-Opitz syndrome bypassing cholesterol: Potential importance of pathway intermediates. Journal of Steroid Biochemistry and Molecular Biology, 2021, 206, 105794.	2.5	12
3	Consensus guidelines for the diagnosis and management of pyridoxineâ€dependent epilepsy due to Î±-amino adipic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 178-192.	3.6	47
4	Deep mining of oxysterols and cholestenoic acids in human plasma and cerebrospinal fluid: Quantification using isotope dilution mass spectrometry. Analytica Chimica Acta, 2021, 1154, 338259.	5.4	14
5	Characterization of Novel Pathogenic Variants Causing Pyridox(am)ine 5â€-Phosphate Oxidase-Dependent Epilepsy. International Journal of Molecular Sciences, 2021, 22, 12013.	4.1	3
6	The effectiveness of correcting abnormal metabolic profiles. Journal of Inherited Metabolic Disease, 2020, 43, 2-13.	3.6	7
7	Rapid, proteomic urine assay for monitoring progressive organ disease in Fabry disease. Journal of Medical Genetics, 2020, 57, 38-47.	3.2	26
8	Organic Solute Transporter Alpha Deficiency: A Disorder With Cholestasis, Liver Fibrosis, and Congenital Diarrhea. Hepatology, 2020, 71, 1879-1882.	7.3	19
9	Is susceptibility to severe COVID-19 disease an inborn error of metabolism?. Journal of Inherited Metabolic Disease, 2020, 43, 906-907.	3.6	8
10	Investigation of diagnostic performance of five urinary cholesterol metabolites for Niemann-Pick disease type C. Journal of Lipid Research, 2019, 60, 2074-2081.	4.2	16
11	Structural Determination of Lysosphingomyelin-509 and Discovery of Novel Class Lipids from Patients with Niemannâ€Pick Disease Type C. International Journal of Molecular Sciences, 2019, 20, 5018.	4.1	25
12	Disorders affecting vitamin B ₆ metabolism. Journal of Inherited Metabolic Disease, 2019, 42, 629-646.	3.6	143
13	Measurement of Bile Acids as a Marker of the Functionality of iPSC-Derived Hepatocytes. Methods in Molecular Biology, 2019, 1994, 141-147.	0.9	0
14	<i>PDXK</i> mutations cause polyneuropathy responsive to pyridoxal 5â€-phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	5.3	54
15	Diagnostic performance evaluation of sulfate-conjugated cholesterol metabolites as urinary biomarkers of Niemannâ€Pick disease type C. Clinica Chimica Acta, 2019, 494, 58-63.	1.1	12
16	Mass Spectrometry Measurement of Albuminâ€Alpha Fetoprotein Ratio as an Indicator of iPSC-Derived Hepatocyte Differentiation. Methods in Molecular Biology, 2019, 1994, 149-156.	0.9	0
17	SNX14 mutations affect endoplasmic reticulum-associated neutral lipid metabolism in autosomal recessive spinocerebellar ataxia 20. Human Molecular Genetics, 2018, 27, 1927-1940.	2.9	71
18	Micronutrients. FIRE Forum for International Research in Education, 2018, 6, 232640981876501.	0.7	0

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19	Sterols and oxysterols in plasma from Smith-Lemli-Opitz syndrome patients. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2017, 169, 77-87.	2.5	34
20	Vps33b is crucial for structural and functional hepatocyte polarity. <i>Journal of Hepatology</i> , 2017, 66, 1001-1011.	3.7	51
21	Quality and stability of extemporaneous pyridoxal phosphate preparations used in the treatment of paediatric epilepsy. <i>Journal of Pharmacy and Pharmacology</i> , 2017, 69, 480-488.	2.4	14
22	Host-Microbe Co-metabolism Dictates Cancer Drug Efficacy in <i>C.Âlegans</i> . <i>Cell</i> , 2017, 169, 442-456.e18.	28.9	198
23	Inherited disorders of transition metal metabolism: an update. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 519-529.	3.6	27
24	An LCâ€MS/MS-Based Method for the Quantification of Pyridox(am)ine 5â€2-Phosphate Oxidase Activity in Dried Blood Spots from Patients with Epilepsy. <i>Analytical Chemistry</i> , 2017, 89, 8892-8900.	6.5	24
25	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. <i>Neurology: Clinical Practice</i> , 2017, 7, 499-511.	1.6	119
26	Mutations in <i>SLC25A22</i> : hyperprolinaemia, vacuolated fibroblasts and presentation with developmental delay. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 385-394.	3.6	16
27	Identification of novel bile acids as biomarkers for the early diagnosis of Niemannâ€Pick C disease. <i>FEBS Letters</i> , 2016, 590, 1651-1662.	2.8	82
28	Mutations in PROSC Disrupt Cellular Pyridoxal Phosphate Homeostasis and Cause Vitamin-B6-Dependent Epilepsy. <i>American Journal of Human Genetics</i> , 2016, 99, 1325-1337.	6.2	118
29	TRNT1 deficiency: clinical, biochemical and molecular genetic features. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 90.	2.7	64
30	Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. <i>Brain</i> , 2016, 139, 2844-2854.	7.6	35
31	<i>RARS</i> mutations in a sibship with infantile spasms. <i>Epilepsia</i> , 2016, 57, e97-e102.	5.1	23
32	<i>ACOX2</i> deficiency: A disorder of bile acid synthesis with transaminase elevation, liver fibrosis, ataxia, and cognitive impairment. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 11289-11293.	7.1	75
33	Mutations in <i>SLC39A14</i> disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€dystonia. <i>Nature Communications</i> , 2016, 7, 11601.	12.8	233
34	Global serum glycoform profiling for the investigation of dystroglycanopathies & Congenital Disorders of Glycosylation. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 7, 55-62.	1.1	8
35	Disorders of Bile Acid Synthesis. , 2016, , 465-475.		3
36	A preterm neonate with seizures unresponsive to conventional treatment. <i>BMJ Case Reports</i> , 2015, 2015, bcr2015209743-bcr2015209743.	0.5	3

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37	Quantitative Charge-Tags for Sterol and Oxysterol Analysis. <i>Clinical Chemistry</i> , 2015, 61, 400-411.	3.2	89
38	Pyridoxamine and pyridoxal are more effective than pyridoxine in rescuing folding-defective variants of human alanine:glyoxylate aminotransferase causing primary hyperoxaluria type I. <i>Human Molecular Genetics</i> , 2015, 24, 5500-5511.	2.9	50
39	Proteomic Discovery and Development of a Multiplexed Targeted MRM-LC-MS/MS Assay for Urine Biomarkers of Extracellular Matrix Disruption in Mucopolysaccharidoses I, II, and VI. <i>Analytical Chemistry</i> , 2015, 87, 12238-12244.	6.5	20
40	Paediatric single mitochondrial DNA deletion disorders: an overlapping spectrum of disease. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 445-457.	3.6	95
41	Normal Cerebrospinal Fluid Pyridoxal 5-Phosphate Level in a PNPO-Deficient Patient with Neonatal-Onset Epileptic Encephalopathy. <i>JIMD Reports</i> , 2015, 22, 67-75.	1.5	21
42	Seizures Due to a KCNQ2 Mutation: Treatment with Vitamin B6. <i>JIMD Reports</i> , 2015, 27, 79-84.	1.5	22
43	Tubular aggregates caused by serine active site containing 1 (SERAC1) mutations in a patient with a mitochondrial encephalopathy. <i>Neuropathology and Applied Neurobiology</i> , 2015, 41, 399-402.	3.2	10
44	Intragenic deletions of ALDH7A1 in pyridoxine-dependent epilepsy caused by Alu - Alu recombination. <i>Neurology</i> , 2015, 85, 756-762.	1.1	34
45	Coenzyme Q10 and Pyridoxal Phosphate Deficiency Is a Common Feature in Mucopolysaccharidosis Type III. <i>JIMD Reports</i> , 2015, 25, 1-7.	1.5	8
46	Treatable childhood neuropathy caused by mutations in riboflavin transporter RFVT2. <i>Brain</i> , 2014, 137, 44-56.	7.6	143
47	Cholestenolic acids regulate motor neuron survival via liver X receptors. <i>Journal of Clinical Investigation</i> , 2014, 124, 4829-4842.	8.2	84
48	Cirrhosis Associated with Pyridoxal 5-Phosphate Treatment of Pyridoxamine 5-Phosphate Oxidase Deficiency. <i>JIMD Reports</i> , 2014, 17, 67-70.	1.5	40
49	Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. <i>Brain</i> , 2014, 137, 1350-1360.	7.6	151
50	Pyridoxine responsiveness in novel mutations of the PNPO gene. <i>Neurology</i> , 2014, 82, 1425-1433.	1.1	100
51	Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. <i>Neurology</i> , 2014, 83, 1873-1875.	1.1	33
52	Liver disease in infancy caused by oxysterol 7- α -hydroxylase deficiency: successful treatment with chenodeoxycholic acid. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 851-861.	3.6	58
53	HIBCH mutations can cause Leigh-like disease with combined deficiency of multiple mitochondrial respiratory chain enzymes and pyruvate dehydrogenase. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 188.	2.7	70
54	Measurement of plasma B ₆ vitamers profiles in children with inborn errors of vitamin B ₆ metabolism using an LC-MS/MS method. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 139-145.	3.6	54

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55	Inborn errors of metabolism causing epilepsy. <i>Developmental Medicine and Child Neurology</i> , 2013, 55, 23-36.	2.1	69
56	Analytical strategies for characterization of oxysterol lipidomes: Liver X receptor ligands in plasma. <i>Free Radical Biology and Medicine</i> , 2013, 59, 69-84.	2.9	56
57	A New Method for the Rapid Diagnosis of Protein N-linked Congenital Disorders of Glycosylation. <i>Journal of Proteome Research</i> , 2013, 12, 3471-3479.	3.7	24
58	Next-Generation Sequencing Reveals Deep Intronic Cryptic ABCC8 and HADH Splicing Founder Mutations Causing Hyperinsulinism by Pseudoexon Activation. <i>American Journal of Human Genetics</i> , 2013, 92, 131-136.	6.2	76
59	MAN1B1 Deficiency: An Unexpected CDG-II. <i>PLoS Genetics</i> , 2013, 9, e1003989.	3.5	63
60	Manganese and the Brain. <i>International Review of Neurobiology</i> , 2013, 110, 277-312.	2.0	159
61	Phenotypic variability in a dystonia family with mutations in the manganese transporter gene. <i>Movement Disorders</i> , 2013, 28, 685-686.	3.9	9
62	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. <i>Brain</i> , 2012, 135, 2875-2882.	7.6	114
63	Urinary AASA excretion is elevated in patients with molybdenum cofactor deficiency and isolated sulphite oxidase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 1031-1036.	3.6	42
64	Dystonia with brain manganese accumulation resulting from <i>SLC30A10</i> mutations: A new treatable disorder. <i>Movement Disorders</i> , 2012, 27, 1317-1322.	3.9	104
65	Differential diagnosis in patients with suspected bile acid synthesis defects. <i>World Journal of Gastroenterology</i> , 2012, 18, 1067.	3.3	38
66	Bile acid-CoA ligase deficiency—a new inborn error of bile acid metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 521-530.	3.6	46
67	Syndrome of Hepatic Cirrhosis, Dystonia, Polycythemia, and Hypermanganesemia Caused by Mutations in <i>SLC30A10</i> , a Manganese Transporter in Man. <i>American Journal of Human Genetics</i> , 2012, 90, 457-466.	6.2	321
68	Disorders of Neurotransmission. , 2012, , 405-422.		3
69	Disorders of Bile Acid Synthesis. , 2012, , 473-484.		0
70	Disorders of Cholesterol Synthesis. , 2012, , 461-471.		1
71	Identification of novel mutations in the proton-coupled folate transporter (PCFT-SLC46A1) associated with hereditary folate malabsorption. <i>Molecular Genetics and Metabolism</i> , 2011, 103, 33-37.	1.1	42
72	Pyridoxine dependent epilepsy and antiquitin deficiency. <i>Molecular Genetics and Metabolism</i> , 2011, 104, 48-60.	1.1	258

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73	The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. <i>Lancet Neurology</i> , The, 2011, 10, 721-733.	10.2	290
74	Disorders of bile acid synthesis. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 593-604.	3.6	134
75	Pyridoxal 5'-phosphate in cerebrospinal fluid; factors affecting concentration. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 529-538.	3.6	68
76	Variable Clinical Spectrum of the Most Common Inborn Error of Bile Acid Metabolism—3-hydroxy- Δ^27 -steroid Dehydrogenase Deficiency. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2010, 50, 61-66.		50
77	An overview of L-2-hydroxyglutarate dehydrogenase gene (L2HGDH) variants: a genotype-phenotype study. <i>Human Mutation</i> , 2010, 31, 380-390.	2.5	108
78	Seizures and paroxysmal events: symptoms pointing to the diagnosis of pyridoxine-dependent epilepsy and pyridoxine phosphate oxidase deficiency. <i>Developmental Medicine and Child Neurology</i> , 2010, 52, e133-42.	2.1	85
79	Pyridoxal 5'-phosphate deficiency causes a loss of aromatic amino acid decarboxylase in patients and human neuroblastoma cells, implications for aromatic amino acid decarboxylase and vitamin B ₆ deficiency states. <i>Journal of Neurochemistry</i> , 2010, 114, 87-96.	3.9	31
80	Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. <i>Brain</i> , 2010, 133, 655-670.	7.6	356
81	Mutations causing Greenberg dysplasia but not Pelger anomaly uncouple enzymatic from structural functions of a nuclear membrane protein. <i>Nucleus</i> , 2010, 1, 354-366.	2.2	44
82	Identification of an unusual variant peroxisome biogenesis disorder caused by mutations in the PEX16 gene. <i>Journal of Medical Genetics</i> , 2010, 47, 608-615.	3.2	80
83	Genotypic and phenotypic spectrum of pyridoxine-dependent epilepsy (ALDH7A1 deficiency). <i>Brain</i> , 2010, 133, 2148-2159.	7.6	219
84	Distal Renal Tubular Acidosis in Filipino Children, Caused by Mutations of the Anion-Exchanger SLC4A1 (AE1, Band 3) Gene. <i>Nephron Physiology</i> , 2010, 114, p19-p24.	1.2	15
85	Genotype-phenotype correlation in PEX5-deficient peroxisome biogenesis defective cell lines. <i>Human Mutation</i> , 2009, 30, 93-98.	2.5	21
86	Analysis of mutant DNA polymerase β in patients with mitochondrial DNA depletion. <i>Human Mutation</i> , 2009, 30, 248-254.	2.5	52
87	Ten novel HMGCL mutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. <i>Human Mutation</i> , 2009, 30, E520-E529.	2.5	21
88	A Nonsense Mutation in COQ9 Causes Autosomal-Recessive Neonatal-Onset Primary Coenzyme Q10 Deficiency: A Potentially Treatable Form of Mitochondrial Disease. <i>American Journal of Human Genetics</i> , 2009, 84, 558-566.	6.2	206
89	Hepatic cirrhosis, dystonia, polycythaemia and hypermanganesaemia—A new metabolic disorder. <i>Journal of Inherited Metabolic Disease</i> , 2008, 31, 151-163.	3.6	114
90	Prenatal testing for a novel EBP missense mutation causing X-linked dominant chondrodysplasia punctata. <i>Prenatal Diagnosis</i> , 2008, 28, 384-388.	2.3	7

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91	Pyridoxal 5â€²-phosphate values in cerebrospinal fluid: Reference values and diagnosis of PNPO deficiency in paediatric patients. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 173-177.	1.1	54
92	Transaldolase deficiency in a two-year-old boy with cirrhosis. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 255-258.	1.1	30
93	Human β -4-3-oxosteroid 5 β -reductase (AKR1D1) deficiency and steroid metabolism. <i>Steroids</i> , 2008, 73, 417-423.	1.8	48
94	Neurotransmitter diseases and related conditions. <i>Molecular Genetics and Metabolism</i> , 2007, 92, 189-197.	1.1	3
95	Mutations in the Gene Encoding 3-Hydroxyisobutyryl-CoA Hydrolase Results in Progressive Infantile Neurodegeneration. <i>American Journal of Human Genetics</i> , 2007, 80, 195-199.	6.2	80
96	An intriguing <i>de novo</i> mutation and a founder effect in <i>antiquitin (ALDH7A1)</i> . <i>Annals of Neurology</i> , 2007, 62, 414-418.	5.3	57
97	Diagnosis of congenital disorders of glycosylation type-I using protein chip technology. <i>Proteomics</i> , 2006, 6, 2295-2304.	2.2	33
98	Mutations in antiquitin in individuals with pyridoxine-dependent seizures. <i>Nature Medicine</i> , 2006, 12, 307-309.	30.7	476
99	B6-responsive disorders: A model of vitamin dependency. <i>Journal of Inherited Metabolic Disease</i> , 2006, 29, 317-326.	3.6	364
100	Disorders of Bile Acid Synthesis. , 2006, , 341-351.		1
101	Disorders of Bile Acid Synthesis. , 2006, , 421-430.		2
102	Disorders of Neurotransmission. , 2006, , 359-372.		0
103	Disorders of Cholesterol Synthesis. , 2006, , 411-420.		0
104	Stomatocytic haemolysis and macrothrombocytopenia (Mediterranean) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 227 Td (stomatocytosis/m British Journal of Haematology, 2005, 130, 297-309.	2.5	138
105	Clinical and genetic spectrum of pyruvate dehydrogenase deficiency: Dihydropyrimidinase (E2) deficiency. <i>Annals of Neurology</i> , 2005, 58, 234-241.	5.3	85
106	Neonatal epileptic encephalopathy caused by mutations in the PNPO gene encoding pyridox(am)ine 5â€²-phosphate oxidase. <i>Human Molecular Genetics</i> , 2005, 14, 1077-1086.	2.9	281
107	Hyperinsulinism of infancy associated with a novel splice site mutation in the SCHAD gene. <i>Journal of Pediatrics</i> , 2005, 146, 706-708.	1.8	68
108	A combined defect in the biosynthesis of N- and O-glycans in patients with cutis laxa and neurological involvement: the biochemical characteristics. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2005, 1741, 156-164.	3.8	34

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109	“African medicine” and Reye's syndrome. <i>Lancet, The</i> , 2004, 363, 860.	13.7	2
110	Mitochondrial HMG-CoA synthase deficiency: identification of two further patients carrying two novel mutations. <i>European Journal of Pediatrics</i> , 2003, 162, 279-280.	2.7	28
111	Novel Mutations in X-Linked Dominant Chondrodysplasia Punctata (CDPX2). <i>Journal of Investigative Dermatology</i> , 2003, 121, 939-942.	0.7	17
112	The underglycosylation of plasma alpha1-antitrypsin in congenital disorders of glycosylation type I is not random. <i>Glycobiology</i> , 2003, 13, 73-85.	2.5	28
113	Correspondence. <i>Pediatric Research</i> , 2003, 53, 865-865.	2.3	1
114	Inborn errors presenting with liver dysfunction. <i>Seminars in Fetal and Neonatal Medicine</i> , 2002, 7, 49-63.	2.7	53
115	Optimisation of Bile Production during Normothermic Preservation of Porcine Livers. <i>American Journal of Transplantation</i> , 2002, 2, 593-599.	4.7	55
116	Synthesis and analysis of conjugates of the major vitamin E metabolite, $\hat{1}\pm$ -CEHC. <i>Free Radical Biology and Medicine</i> , 2002, 33, 807-817.	2.9	63
117	Neonatal presentation of coenzyme Q10 deficiency. <i>Journal of Pediatrics</i> , 2001, 139, 456-458.	1.8	112
118	Diversity of congenital disorders of glycosylation. <i>Lancet, The</i> , 2001, 357, 1382-1383.	13.7	22
119	Identification of $\hat{1}\pm$ 1-Antitrypsin Variants in Plasma with the Use of Proteomic Technology. <i>Clinical Chemistry</i> , 2001, 47, 2012-2022.	3.2	52
120	Congenital disorders of glycosylation type I leads to altered processing of N-linked glycans, as well as underglycosylation. <i>Biochemical Journal</i> , 2001, 359, 249.	3.7	21
121	Analysis by matrix assisted laser desorption/ ionisation-time of flight mass spectrometry of the post-translational modifications of $\hat{1}\pm$ 1-antitrypsin isoforms separated by two-dimensional polyacrylamide gel electrophoresis. <i>Proteomics</i> , 2001, 1, 778-786.	2.2	44
122	New synthesis of ($\hat{A}\pm$)- $\hat{1}\pm$ -CMBHC and its confirmation as a metabolite of $\hat{1}\pm$ -tocopherol (vitamin E). <i>Biorganic and Medicinal Chemistry</i> , 2001, 9, 1337-1343.	3.0	23
123	Hyperinsulinism in short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency reveals the importance of $\hat{1}^2$ -oxidation in insulin secretion. <i>Journal of Clinical Investigation</i> , 2001, 108, 457-465.	8.2	246
124	A strategy for the identification of site-specific glycosylation in glycoproteins using MALDI TOF MS. <i>Tetrahedron: Asymmetry</i> , 2000, 11, 75-93.	1.8	21
125	Mutations in the gene encoding peroxisomal $\hat{1}\pm$ -methylacyl-CoA racemase cause adult-onset sensory motor neuropathy. <i>Nature Genetics</i> , 2000, 24, 188-191.	21.4	241
126	Effect of Intravenous Lipid Emulsions on Hepatic Cholesterol Metabolism. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2000, 30, 538-546.	1.8	14

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127	The identification of unusual bile acid metabolites by tandem mass spectrometry: use of low-energy collision-induced dissociation to produce informative spectra. , 1999, 13, 1159-1164.		18
128	The Role of Phytosterols in the Pathogenesis of Liver Complications of Pediatric Parenteral Nutrition. Nutrition, 1998, 14, 158-164.	2.4	213
129	Prenatal diagnosis of the carbohydrate-deficient glycoprotein syndrome type 1A (CDG1A) by a combination of enzymology and genetic linkage analysis after amniocentesis or chorionic villus sampling. , 1998, 18, 693-699.		21
130	Clinical phenotype of desmosterolosis. , 1998, 75, 145-152.		157
131	Infantile parkinsonism-dystonia: Tyrosine hydroxylase deficiency. Movement Disorders, 1998, 13, 350-350.	3.9	12
132	Alstr�m syndrome. Ophthalmology, 1998, 105, 1274-1280.	5.2	150
133	Liver failure associated with mitochondrial DNA depletion. Journal of Hepatology, 1998, 28, 556-563.	3.7	106
134	Defective galactosylation of serum transferrin in galactosemia. Glycobiology, 1998, 8, 351-357.	2.5	115
135	Clinical phenotype of desmosterolosis. American Journal of Medical Genetics Part A, 1998, 75, 145-152.	2.4	4
136	A Method for the Quantitation of Conjugated Bile Acids in Dried Blood Spots Using Electrospray Ionization-Mass Spectrometry. Pediatric Research, 1998, 43, 361-368.	2.3	51
137	Desmosterolosis: a new inborn error of cholesterol biosynthesis. Lancet, The, 1996, 348, 404.	13.7	55
138	Recessively inherited L-DOPA-responsive parkinsonism in infancy caused by a point mutation (L205P) in the tyrosine hydroxylase gene. Human Molecular Genetics, 1996, 5, 1023-1028.	2.9	175
139	Sphincterotomy for Jaundice in a Neonate. Journal of Pediatric Gastroenterology and Nutrition, 1996, 23, 507-509.	1.8	11
140	First Trimester Prenatal Diagnosis of Smith-Lemli-Opitz Syndrome(7-Dehydrocholesterol Reductase) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	2.3	42
141	Concerning "Agamanolis disease", American Journal of Medical Genetics Part A, 1995, 56, 289-289.	2.4	2
142	Increased first trimester nuchal translucency as a prenatal manifestation of Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics Part A, 1995, 58, 374-376.	2.4	59