Peter Theodore Clayton

List of Publications by Year
in descending order

142 papers	10,009 citations	29994 54 h-index	39575 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 Ï‰-Oxidation. International Journal of Molecular Sciences, 2022, 23, 987.	1.8	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.	1.2	12
3	Consensus guidelines for the diagnosis and management of pyridoxineâ€dependent epilepsy due to Î±â€aminoadipic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 178-192.	1.7	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.	2.6	14
5	Characterization of Novel Pathogenic Variants Causing Pyridox(am)ine 5â€²-Phosphate Oxidase-Dependent Epilepsy. International Journal of Molecular Sciences, 2021, 22, 12013.	1.8	3
6	The effectiveness of correcting abnormal metabolic profiles. Journal of Inherited Metabolic Disease, 2020, 43, 2-13.	1.7	7
7	Rapid, proteomic urine assay for monitoring progressive organ disease in Fabry disease. Journal of Medical Genetics, 2020, 57, 38-47.	1.5	26
8	Organic Solute Transporter Alpha Deficiency: A Disorder With Cholestasis, Liver Fibrosis, and Congenital Diarrhea. Hepatology, 2020, 71, 1879-1882.	3.6	19
9	Is susceptibility to severe <scp>COVID</scp>â€19 disease an inborn error of metabolism?. Journal of Inherited Metabolic Disease, 2020, 43, 906-907.	1.7	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.	2.0	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.	1.8	25
12	Disorders affecting vitamin B₆ metabolism. Journal of Inherited Metabolic Disease, 2019, 42, 629-646.	1.7	143
13	Measurement of Bile Acids as a Marker of the Functionality of iPSC-Derived Hepatocytes. Methods in Molecular Biology, 2019, 1994, 141-147.	0.4	0
14	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	2.8	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.	0.5	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.	0.4	0
17	SNX14 mutations affect endoplasmic reticulum-associated neutral lipid metabolism in autosomal recessive spinocerebellar ataxia 20. Human Molecular Genetics, 2018, 27, 1927-1940.	1.4	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. Journal of Steroid Biochemistry and Molecular Biology, 2017, 169, 77-87.	1.2	34
20	Vps33b is crucial for structural and functional hepatocyte polarity. Journal of Hepatology, 2017, 66, 1001-1011.	1.8	51
21	Quality and stability of extemporaneous pyridoxal phosphate preparations used in the treatment of paediatric epilepsy. Journal of Pharmacy and Pharmacology, 2017, 69, 480-488.	1.2	14
22	Host-Microbe Co-metabolism Dictates Cancer Drug Efficacy in C.Âelegans. Cell, 2017, 169, 442-456.e18.	13.5	198
23	Inherited disorders of transition metal metabolism: an update. Journal of Inherited Metabolic Disease, 2017, 40, 519-529.	1.7	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. Analytical Chemistry, 2017, 89, 8892-8900.	3.2	24
25	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. Neurology: Clinical Practice, 2017, 7, 499-511.	0.8	119
26	Mutations in SLC25A22: hyperprolinaemia, vacuolated fibroblasts and presentation with developmental delay. Journal of Inherited Metabolic Disease, 2017, 40, 385-394.	1.7	16
27	Identification of novel bile acids as biomarkers for the early diagnosis of Niemannâ€Pick C disease. FEBS Letters, 2016, 590, 1651-1662.	1.3	82
28	Mutations in PROSC Disrupt Cellular Pyridoxal Phosphate Homeostasis and Cause Vitamin-B6-Dependent Epilepsy. American Journal of Human Genetics, 2016, 99, 1325-1337.	2.6	118
29	TRNT1 deficiency: clinical, biochemical and molecular genetic features. Orphanet Journal of Rare Diseases, 2016, 11, 90.	1.2	64
30	Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. Brain, 2016, 139, 2844-2854.	3.7	35
31	<scp>RARS</scp>2 mutations in a sibship with infantile spasms. Epilepsia, 2016, 57, e97-e102.	2.6	23
32	ACOX2 deficiency: A disorder of bile acid synthesis with transaminase elevation, liver fibrosis, ataxia, and cognitive impairment. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 11289-11293.	3.3	75
33	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€“dystonia. Nature Communications, 2016, 7, 11601.	5.8	233
34	Global serum glycoform profiling for the investigation of dystroglycanopathies & Congenital Disorders of Glycosylation. Molecular Genetics and Metabolism Reports, 2016, 7, 55-62.	0.4	8
35	Disorders of Bile Acid Synthesis. , 2016, , 465-475.		3
36	A preterm neonate with seizures unresponsive to conventional treatment. BMJ Case Reports, 2015, 2015, bcr2015209743-bcr2015209743.	0.2	3

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37	Quantitative Charge-Tags for Sterol and Oxysterol Analysis. Clinical Chemistry, 2015, 61, 400-411.	1.5	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. Human Molecular Genetics, 2015, 24, 5500-5511.	1.4	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. Analytical Chemistry, 2015, 87, 12238-12244.	3.2	20
40	Paediatric single mitochondrial DNA deletion disorders: an overlapping spectrum of disease. Journal of Inherited Metabolic Disease, 2015, 38, 445-457.	1.7	95
41	Normal Cerebrospinal Fluid Pyridoxal 5â€²-Phosphate Level in a PNPO-Deficient Patient with Neonatal-Onset Epileptic Encephalopathy. JIMD Reports, 2015, 22, 67-75.	0.7	21
42	Seizures Due to a KCNQ2 Mutation: Treatment with Vitamin B6. JIMD Reports, 2015, 27, 79-84.	0.7	22
43	Tubular aggregates caused by serine active site containing 1 (<scp>SERAC1</scp>) mutations in a patient with a mitochondrial encephalopathy. Neuropathology and Applied Neurobiology, 2015, 41, 399-402.	1.8	10
44	Intragenic deletions of ALDH7A1 in pyridoxine-dependent epilepsy caused by Alu - Alu recombination. Neurology, 2015, 85, 756-762.	1.5	34
45	Coenzyme Q10 and Pyridoxal Phosphate Deficiency Is a Common Feature in Mucopolysaccharidosis Type III. JIMD Reports, 2015, 25, 1-7.	0.7	8
46	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. Brain, 2014, 137, 44-56.	3.7	143
47	Cholestenoic acids regulate motor neuron survival via liver X receptors. Journal of Clinical Investigation, 2014, 124, 4829-4842.	3.9	84
48	Cirrhosis Associated with Pyridoxal 5â€²-Phosphate Treatment of Pyridoxamine 5â€²-Phosphate Oxidase Deficiency. JIMD Reports, 2014, 17, 67-70.	0.7	40
49	Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. Brain, 2014, 137, 1350-1360.	3.7	151
50	Pyridoxine responsiveness in novel mutations of the PNPO gene. Neurology, 2014, 82, 1425-1433.	1.5	100
51	Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. Neurology, 2014, 83, 1873-1875.	1.5	33
52	Liver disease in infancy caused by oxysterol 7Î±â€hydroxylase deficiency: successful treatment with chenodeoxycholic acid. Journal of Inherited Metabolic Disease, 2014, 37, 851-861.	1.7	58
53	HIBCH mutations can cause Leigh-like disease with combined deficiency of multiple mitochondrial respiratory chain enzymes and pyruvate dehydrogenase. Orphanet Journal of Rare Diseases, 2013, 8, 188.	1.2	70
54	Measurement of plasma B₆ vitamer profiles in children with inborn errors of vitamin B₆ metabolism using an LCâ€MS/MS method. Journal of Inherited Metabolic Disease, 2013, 36, 139-145.	1.7	54

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55	Inborn errors of metabolism causing epilepsy. Developmental Medicine and Child Neurology, 2013, 55, 23-36.	1.1	69
56	Analytical strategies for characterization of oxysterol lipidomes: Liver X receptor ligands in plasma. Free Radical Biology and Medicine, 2013, 59, 69-84.	1.3	56
57	A New Method for the Rapid Diagnosis of Protein N-linked Congenital Disorders of Glycosylation. Journal of Proteome Research, 2013, 12, 3471-3479.	1.8	24
58	Next-Generation Sequencing Reveals Deep Intronic Cryptic ABCC8 and HADH Splicing Founder Mutations Causing Hyperinsulinism by Pseudoexon Activation. American Journal of Human Genetics, 2013, 92, 131-136.	2.6	76
59	MAN1B1 Deficiency: An Unexpected CDG-II. PLoS Genetics, 2013, 9, e1003989.	1.5	63
60	Manganese and the Brain. International Review of Neurobiology, 2013, 110, 277-312.	0.9	159
61	Phenotypic variability in a dystonia family with mutations in the manganese transporter gene. Movement Disorders, 2013, 28, 685-686.	2.2	9
62	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. Brain, 2012, 135, 2875-2882.	3.7	114
63	Urinary AASA excretion is elevated in patients with molybdenum cofactor deficiency and isolated sulphite oxidase deficiency. Journal of Inherited Metabolic Disease, 2012, 35, 1031-1036.	1.7	42
64	Dystonia with brain manganese accumulation resulting from SLC30A10 mutations: A new treatable disorder. Movement Disorders, 2012, 27, 1317-1322.	2.2	104
65	Differential diagnosis in patients with suspected bile acid synthesis defects. World Journal of Gastroenterology, 2012, 18, 1067.	1.4	38
66	Bile acidâ€CoA ligase deficiencyâ€"a new inborn error of bile acid metabolism. Journal of Inherited Metabolic Disease, 2012, 35, 521-530.	1.7	46
67	Syndrome of Hepatic Cirrhosis, Dystonia, Polycythemia, and Hypermanganesemia Caused by Mutations in SLC30A10 , a Manganese Transporter in Man. American Journal of Human Genetics, 2012, 90, 457-466.	2.6	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. Molecular Genetics and Metabolism, 2011, 103, 33-37.	0.5	42
72	Pyridoxine dependent epilepsy and antiquitin deficiency. Molecular Genetics and Metabolism, 2011, 104, 48-60.	0.5	258

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73	The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. Lancet Neurology, The, 2011, 10, 721-733.	4.9	290
74	Disorders of bile acid synthesis. Journal of Inherited Metabolic Disease, 2011, 34, 593-604.	1.7	134
75	Pyridoxal 5'â€phosphate in cerebrospinal fluid; factors affecting concentration. Journal of Inherited Metabolic Disease, 2011, 34, 529-538.	1.7	68
76	Variable Clinical Spectrum of the Most Common Inborn Error of Bile Acid Metabolismâ€"3Î²â€hydroxyâ€Î”⁵â€C₂₇â€steroid Dehydrogenase Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2010, 50, 61-66.	0.9	50
77	An overview of L-2-hydroxyglutarate dehydrogenase gene (L2HGDH) variants: a genotype-phenotype study. Human Mutation, 2010, 31, 380-390.	1.1	108
78	Seizures and paroxysmal events: symptoms pointing to the diagnosis of pyridoxineâ€dependent epilepsy and pyridoxine phosphate oxidase deficiency. Developmental Medicine and Child Neurology, 2010, 52, e133-42.	1.1	85
79	Pyridoxal 5â€²â€phosphate deficiency causes a loss of aromatic lâ€amino acid decarboxylase in patients and human neuroblastoma cells, implications for aromatic lâ€amino acid decarboxylase and vitamin B₆ deficiency states. Journal of Neurochemistry, 2010, 114, 87-96.	2.1	31
80	Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. Brain, 2010, 133, 655-670.	3.7	356
81	Mutations causing Greenberg dysplasia but not Pelger anomaly uncouple enzymatic from structural functions of a nuclear membrane protein. Nucleus, 2010, 1, 354-366.	0.6	44
82	Identification of an unusual variant peroxisome biogenesis disorder caused by mutations in the PEX16 gene. Journal of Medical Genetics, 2010, 47, 608-615.	1.5	80
83	Genotypic and phenotypic spectrum of pyridoxine-dependent epilepsy (ALDH7A1 deficiency). Brain, 2010, 133, 2148-2159.	3.7	219
84	Distal Renal Tubular Acidosis in Filipino Children, Caused by Mutations of the Anion-Exchanger SLC4A1 (AE1, Band 3) Gene. Nephron Physiology, 2010, 114, p19-p24.	1.5	15
85	Genotype-phenotype correlation in PEX5-deficient peroxisome biogenesis defective cell lines. Human Mutation, 2009, 30, 93-98.	1.1	21
86	Analysis of mutant DNA polymerase Î³ in patients with mitochondrial DNA depletion. Human Mutation, 2009, 30, 248-254.	1.1	52
87	Ten novelHMGCLmutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. Human Mutation, 2009, 30, E520-E529.	1.1	21
88	A Nonsense Mutation in COQ9 Causes Autosomal-Recessive Neonatal-Onset Primary Coenzyme Q10 Deficiency: A Potentially Treatable Form of Mitochondrial Disease. American Journal of Human Genetics, 2009, 84, 558-566.	2.6	206
89	Hepatic cirrhosis, dystonia, polycythaemia and hypermanganesaemiaâ€"A new metabolic disorder. Journal of Inherited Metabolic Disease, 2008, 31, 151-163.	1.7	114
90	Prenatal testing for a novelEBP missense mutation causing X-linked dominant chondrodysplasia punctata. Prenatal Diagnosis, 2008, 28, 384-388.	1.1	7

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91	Pyridoxal 5â€²-phosphate values in cerebrospinal fluid: Reference values and diagnosis of PNPO deficiency in paediatric patients. Molecular Genetics and Metabolism, 2008, 94, 173-177.	0.5	54
92	Transaldolase deficiency in a two-year-old boy with cirrhosis. Molecular Genetics and Metabolism, 2008, 94, 255-258.	0.5	30
93	Human Î”4-3-oxosteroid 5Î²-reductase (AKR1D1) deficiency and steroid metabolism. Steroids, 2008, 73, 417-423.	0.8	48
94	Neurotransmitter diseases and related conditions. Molecular Genetics and Metabolism, 2007, 92, 189-197.	0.5	3
95	Mutations in the Gene Encoding 3-Hydroxyisobutyryl-CoA Hydrolase Results in Progressive Infantile Neurodegeneration. American Journal of Human Genetics, 2007, 80, 195-199.	2.6	80
96	An intriguing â€œsilentâ€•mutation and a founder effect in antiquitin (ALDH7A1). Annals of Neurology, 2007, 62, 414-418.	2.8	57
97	Diagnosis of congenital disorders of glycosylation type-I using protein chip technology. Proteomics, 2006, 6, 2295-2304.	1.3	33
98	Mutations in antiquitin in individuals with pyridoxine-dependent seizures. Nature Medicine, 2006, 12, 307-309.	15.2	476
99	B6-responsive disorders: A model of vitamin dependency. Journal of Inherited Metabolic Disease, 2006, 29, 317-326.	1.7	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.	1.2	138
105	Clinical and genetic spectrum of pyruvate dehydrogenase deficiency: Dihydrolipoamide acetyltransferase (E2) deficiency. Annals of Neurology, 2005, 58, 234-241.	2.8	85
106	Neonatal epileptic encephalopathy caused by mutations in the PNPO gene encoding pyridox(am)ine 5â€²-phosphate oxidase. Human Molecular Genetics, 2005, 14, 1077-1086.	1.4	281
107	Hyperinsulinism of infancy associated with a novel splice site mutation in the SCHAD gene. Journal of Pediatrics, 2005, 146, 706-708.	0.9	68
108	A combined defect in the biosynthesis of N- and O-glycans in patients with cutis laxa and neurological involvement: the biochemical characteristics. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2005, 1741, 156-164.	1.8	34

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109	â€œAfrican medicineâ€•and Reye's syndrome. Lancet, The, 2004, 363, 860.	6.3	2
110	Mitochondrial HMG-CoA synthase deficiency: identification of two further patients carrying two novel mutations. European Journal of Pediatrics, 2003, 162, 279-280.	1.3	28
111	Novel Mutations in X-Linked Dominant Chondrodysplasia Punctata (CDPX2). Journal of Investigative Dermatology, 2003, 121, 939-942.	0.3	17
112	The underglycosylation of plasma alpha1-antitrypsin in congenital disorders of glycosylation type I is not random. Glycobiology, 2003, 13, 73-85.	1.3	28
113	Correspondence. Pediatric Research, 2003, 53, 865-865.	1.1	1
114	Inborn errors presenting with liver dysfunction. Seminars in Fetal and Neonatal Medicine, 2002, 7, 49-63.	2.8	53
115	Optimisation of Bile Production during Normothermic Preservation of Porcine Livers. American Journal of Transplantation, 2002, 2, 593-599.	2.6	55
116	Synthesis and analysis of conjugates of the major vitamin E metabolite, Î±-CEHC. Free Radical Biology and Medicine, 2002, 33, 807-817.	1.3	63
117	Neonatal presentation of coenzyme Q10 deficiency. Journal of Pediatrics, 2001, 139, 456-458.	0.9	112
118	Diversity of congenital disorders of glycosylation. Lancet, The, 2001, 357, 1382-1383.	6.3	22
119	Identification of Î±1-Antitrypsin Variants in Plasma with the Use of Proteomic Technology. Clinical Chemistry, 2001, 47, 2012-2022.	1.5	52
120	Congenital disorders of glycosylation type I leads to altered processing of N-linked glycans, as well as underglycosylation. Biochemical Journal, 2001, 359, 249.	1.7	21
121	Analysis by matrix assisted laser desorption/ ionisation-time of flight mass spectrometry of the post-translational modifications of Î±1-antitrypsin isoforms separated by two- dimensional polyacrylamide gel electrophoresis. Proteomics, 2001, 1, 778-786.	1.3	44
122	New synthesis of (Â±)-Î±-CMBHC and its confirmation as a metabolite of Î±-tocopherol (vitamin E). Bioorganic and Medicinal Chemistry, 2001, 9, 1337-1343.	1.4	23
123	Hyperinsulinism in short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency reveals the importance of Î²-oxidation in insulin secretion. Journal of Clinical Investigation, 2001, 108, 457-465.	3.9	246
124	A strategy for the identification of site-specific glycosylation in glycoproteins using MALDI TOF MS. Tetrahedron: Asymmetry, 2000, 11, 75-93.	1.8	21
125	Mutations in the gene encoding peroxisomal Î±-methylacyl-CoA racemase cause adult-onset sensory motor neuropathy. Nature Genetics, 2000, 24, 188-191.	9.4	241
126	Effect of Intravenous Lipid Emulsions on Hepatic Cholesterol Metabolism. Journal of Pediatric Gastroenterology and Nutrition, 2000, 30, 538-546.	0.9	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.	1.1	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.	2.2	12
132	AlstrÃ¶m syndrome. Ophthalmology, 1998, 105, 1274-1280.	2.5	150
133	Liver failure associated with mitochondrial DNA depletion. Journal of Hepatology, 1998, 28, 556-563.	1.8	106
134	Defective galactosylation of serum transferrin in galactosemia. Glycobiology, 1998, 8, 351-357.	1.3	115
135	Clinical phenotype of desmosterolosis. , 1998, 75, 145.		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.	1.1	51
137	Desmosterolosis: a new inborn error of cholesterol biosynthesis. Lancet, The, 1996, 348, 404.	6.3	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.	1.4	175
139	Sphincterotomy for Jaundice in a Neonate. Journal of Pediatric Gastroenterology and Nutrition, 1996, 23, 507-509.	0.9	11
140	First Trimester Prenatal Diagnosis of Smith-Lemli-Opitz Syndrome(7-Dehydrocholesterol Reductase) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	1.1	42
141	Concerning â€œAgamanolis diseaseâ€• 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