

# Peter Theodore Clayton

## List of Publications by Year in descending order

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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	Mutations in antiquitin in individuals with pyridoxine-dependent seizures. <i>Nature Medicine</i> , 2006, 12, 307-309.	15.2	476
2	B6-responsive disorders: A model of vitamin dependency. <i>Journal of Inherited Metabolic Disease</i> , 2006, 29, 317-326.	1.7	364
3	Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. <i>Brain</i> , 2010, 133, 655-670.	3.7	356
4	Syndrome of Hepatic Cirrhosis, Dystonia, Polycythemia, and Hypermanganesemia Caused by Mutations in SLC30A10, a Manganese Transporter in Man. <i>American Journal of Human Genetics</i> , 2012, 90, 457-466.	2.6	321
5	The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. <i>Lancet Neurology</i> , The, 2011, 10, 721-733.	4.9	290
6	Neonatal epileptic encephalopathy caused by mutations in the PNPO gene encoding pyridox(am)ine 5 $\alpha$ -phosphate oxidase. <i>Human Molecular Genetics</i> , 2005, 14, 1077-1086.	1.4	281
7	Pyridoxine dependent epilepsy and antiquitin deficiency. <i>Molecular Genetics and Metabolism</i> , 2011, 104, 48-60.	0.5	258
8	Hyperinsulinism in short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency reveals the importance of $\beta$ -oxidation in insulin secretion. <i>Journal of Clinical Investigation</i> , 2001, 108, 457-465.	3.9	246
9	Mutations in the gene encoding peroxisomal $\beta$ -methylacyl-CoA racemase cause adult-onset sensory motor neuropathy. <i>Nature Genetics</i> , 2000, 24, 188-191.	9.4	241
10	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonism and dystonia. <i>Nature Communications</i> , 2016, 7, 11601.	5.8	233
11	Genotypic and phenotypic spectrum of pyridoxine-dependent epilepsy (ALDH7A1 deficiency). <i>Brain</i> , 2010, 133, 2148-2159.	3.7	219
12	The Role of Phytosterols in the Pathogenesis of Liver Complications of Pediatric Parenteral Nutrition. <i>Nutrition</i> , 1998, 14, 158-164.	1.1	213
13	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.	2.6	206
14	Host-Microbe Co-metabolism Dictates Cancer Drug Efficacy in <i>C. elegans</i> . <i>Cell</i> , 2017, 169, 442-456.e18.	13.5	198
15	Recessively inherited L-DOPA-responsive parkinsonism in infancy caused by a point mutation (L205P) in the tyrosine hydroxylase gene. <i>Human Molecular Genetics</i> , 1996, 5, 1023-1028.	1.4	175
16	Manganese and the Brain. <i>International Review of Neurobiology</i> , 2013, 110, 277-312.	0.9	159
17	Clinical phenotype of desmosterolosis. , 1998, 75, 145-152.		157
18	Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. <i>Brain</i> , 2014, 137, 1350-1360.	3.7	151

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19	Alström syndrome. <i>Ophthalmology</i> , 1998, 105, 1274-1280.	2.5	150
20	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. <i>Brain</i> , 2014, 137, 44-56.	3.7	143
21	Disorders affecting vitamin B <sub>6</sub> metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 629-646.	1.7	143
22	Stomatocytic haemolysis and macrothrombocytopenia (Mediterranean) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 627 Td (stomatocytosis/m British Journal of Haematology, 2005, 130, 297-309.	1.2	138
23	Disorders of bile acid synthesis. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 593-604.	1.7	134
24	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. <i>Neurology: Clinical Practice</i> , 2017, 7, 499-511.	0.8	119
25	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.	2.6	118
26	Defective galactosylation of serum transferrin in galactosemia. <i>Glycobiology</i> , 1998, 8, 351-357.	1.3	115
27	Hepatic cirrhosis, dystonia, polycythaemia and hypermanganesaemia—A new metabolic disorder. <i>Journal of Inherited Metabolic Disease</i> , 2008, 31, 151-163.	1.7	114
28	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. <i>Brain</i> , 2012, 135, 2875-2882.	3.7	114
29	Neonatal presentation of coenzyme Q10 deficiency. <i>Journal of Pediatrics</i> , 2001, 139, 456-458.	0.9	112
30	An overview of L-2-hydroxyglutarate dehydrogenase gene (L2HGDH) variants: a genotype-phenotype study. <i>Human Mutation</i> , 2010, 31, 380-390.	1.1	108
31	Liver failure associated with mitochondrial DNA depletion. <i>Journal of Hepatology</i> , 1998, 28, 556-563.	1.8	106
32	Dystonia with brain manganese accumulation resulting from <i>SLC30A10</i> mutations: A new treatable disorder. <i>Movement Disorders</i> , 2012, 27, 1317-1322.	2.2	104
33	Pyridoxine responsiveness in novel mutations of the <i>PNPO</i> gene. <i>Neurology</i> , 2014, 82, 1425-1433.	1.5	100
34	Paediatric single mitochondrial DNA deletion disorders: an overlapping spectrum of disease. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 445-457.	1.7	95
35	Quantitative Charge-Tags for Sterol and Oxysterol Analysis. <i>Clinical Chemistry</i> , 2015, 61, 400-411.	1.5	89
36	Clinical and genetic spectrum of pyruvate dehydrogenase deficiency: Dihydrolipoamide acetyltransferase (E2) deficiency. <i>Annals of Neurology</i> , 2005, 58, 234-241.	2.8	85

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37	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.	1.1	85
38	Cholestenolic acids regulate motor neuron survival via liver X receptors. <i>Journal of Clinical Investigation</i> , 2014, 124, 4829-4842.	3.9	84
39	Identification of novel bile acids as biomarkers for the early diagnosis of Niemann-Pick C disease. <i>FEBS Letters</i> , 2016, 590, 1651-1662.	1.3	82
40	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.	2.6	80
41	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.	1.5	80
42	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.	2.6	76
43	<i>ACO2</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.	3.3	75
44	SNX14 mutations affect endoplasmic reticulum-associated neutral lipid metabolism in autosomal recessive spinocerebellar ataxia 20. <i>Human Molecular Genetics</i> , 2018, 27, 1927-1940.	1.4	71
45	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.	1.2	70
46	Inborn errors of metabolism causing epilepsy. <i>Developmental Medicine and Child Neurology</i> , 2013, 55, 23-36.	1.1	69
47	Hyperinsulinism of infancy associated with a novel splice site mutation in the SCHAD gene. <i>Journal of Pediatrics</i> , 2005, 146, 706-708.	0.9	68
48	Pyridoxal 5'-phosphate in cerebrospinal fluid; factors affecting concentration. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 529-538.	1.7	68
49	TRNT1 deficiency: clinical, biochemical and molecular genetic features. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 90.	1.2	64
50	Synthesis and analysis of conjugates of the major vitamin E metabolite, $\alpha$ -CEHC. <i>Free Radical Biology and Medicine</i> , 2002, 33, 807-817.	1.3	63
51	MAN1B1 Deficiency: An Unexpected CDG-II. <i>PLoS Genetics</i> , 2013, 9, e1003989.	1.5	63
52	Increased first trimester nuchal translucency as a prenatal manifestation of Smith-Lemli-Opitz syndrome. <i>American Journal of Medical Genetics Part A</i> , 1995, 58, 374-376.	2.4	59
53	Liver disease in infancy caused by oxysterol $7\alpha$ -hydroxylase deficiency: successful treatment with chenodeoxycholic acid. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 851-861.	1.7	58
54	An intriguing "silent" mutation and a founder effect in <i>antiquitin (ALDH7A1)</i> . <i>Annals of Neurology</i> , 2007, 62, 414-418.	2.8	57

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55	Analytical strategies for characterization of oxysterol lipidomes: Liver X receptor ligands in plasma. <i>Free Radical Biology and Medicine</i> , 2013, 59, 69-84.	1.3	56
56	Desmosterolosis: a new inborn error of cholesterol biosynthesis. <i>Lancet</i> , The, 1996, 348, 404.	6.3	55
57	Optimisation of Bile Production during Normothermic Preservation of Porcine Livers. <i>American Journal of Transplantation</i> , 2002, 2, 593-599.	2.6	55
58	Pyridoxal 5-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.	0.5	54
59	Measurement of plasma B <sub>6</sub> vitamers profiles in children with inborn errors of vitamin B <sub>6</sub> metabolism using an LC-MS/MS method. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 139-145.	1.7	54
60	<i>PDXK</i> mutations cause polyneuropathy responsive to pyridoxal 5-phosphate supplementation. <i>Annals of Neurology</i> , 2019, 86, 225-240.	2.8	54
61	Inborn errors presenting with liver dysfunction. <i>Seminars in Fetal and Neonatal Medicine</i> , 2002, 7, 49-63.	2.8	53
62	Identification of $\pm$ 1-Antitrypsin Variants in Plasma with the Use of Proteomic Technology. <i>Clinical Chemistry</i> , 2001, 47, 2012-2022.	1.5	52
63	Analysis of mutant DNA polymerase $\beta$ in patients with mitochondrial DNA depletion. <i>Human Mutation</i> , 2009, 30, 248-254.	1.1	52
64	Vps33b is crucial for structural and functional hepatocyte polarity. <i>Journal of Hepatology</i> , 2017, 66, 1001-1011.	1.8	51
65	A Method for the Quantitation of Conjugated Bile Acids in Dried Blood Spots Using Electrospray Ionization-Mass Spectrometry. <i>Pediatric Research</i> , 1998, 43, 361-368.	1.1	51
66	Variable Clinical Spectrum of the Most Common Inborn Error of Bile Acid Metabolism: 3-hydroxy- $\Delta^5$ - $\Delta^27$ -steroid Dehydrogenase Deficiency. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2010, 50, 61-66.	0.9	50
67	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.	1.4	50
68	Human $\Delta^4$ -3-oxosteroid 5-reductase (AKR1D1) deficiency and steroid metabolism. <i>Steroids</i> , 2008, 73, 417-423.	0.8	48
69	Consensus guidelines for the diagnosis and management of pyridoxine-dependent epilepsy due to $\pm$ -amino adipic semialdehyde dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 178-192.	1.7	47
70	Bile acid-CoA ligase deficiency: a new inborn error of bile acid metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 521-530.	1.7	46
71	Analysis by matrix assisted laser desorption/ionisation-time of flight mass spectrometry of the post-translational modifications of $\pm$ 1-antitrypsin isoforms separated by two-dimensional polyacrylamide gel electrophoresis. <i>Proteomics</i> , 2001, 1, 778-786.	1.3	44
72	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.	0.6	44

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73	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.	0.5	42
74	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.	1.7	42
75	First Trimester Prenatal Diagnosis of Smith-Lemli-Opitz Syndrome(7-Dehydrocholesterol Reductase) Tj ETQq1 1 0.784314 rgBT/Overl 1.1	1.1	42
76	Cirrhosis Associated with Pyridoxal 5â€²-Phosphate Treatment of Pyridoxamine 5â€²-Phosphate Oxidase Deficiency. <i>JIMD Reports</i> , 2014, 17, 67-70.	0.7	40
77	Differential diagnosis in patients with suspected bile acid synthesis defects. <i>World Journal of Gastroenterology</i> , 2012, 18, 1067.	1.4	38
78	Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. <i>Brain</i> , 2016, 139, 2844-2854.	3.7	35
79	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.	1.8	34
80	Intragenic deletions of <i>ALDH7A1</i> in pyridoxine-dependent epilepsy caused by <i>Alu</i> - <i>Alu</i> recombination. <i>Neurology</i> , 2015, 85, 756-762.	1.5	34
81	Sterols and oxysterols in plasma from Smith-Lemli-Opitz syndrome patients. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2017, 169, 77-87.	1.2	34
82	Diagnosis of congenital disorders of glycosylation type-I using protein chip technology. <i>Proteomics</i> , 2006, 6, 2295-2304.	1.3	33
83	Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. <i>Neurology</i> , 2014, 83, 1873-1875.	1.5	33
84	Pyridoxal 5â€²-â€²phosphate deficiency causes a loss of aromatic lâ€²-amino acid decarboxylase in patients and human neuroblastoma cells, implications for aromatic lâ€²-amino acid decarboxylase and vitamin B<sub>6</sub> deficiency states. <i>Journal of Neurochemistry</i> , 2010, 114, 87-96.	2.1	31
85	Transaldolase deficiency in a two-year-old boy with cirrhosis. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 255-258.	0.5	30
86	Mitochondrial HMG-CoA synthase deficiency: identification of two further patients carrying two novel mutations. <i>European Journal of Pediatrics</i> , 2003, 162, 279-280.	1.3	28
87	The underglycosylation of plasma alpha1-antitrypsin in congenital disorders of glycosylation type I is not random. <i>Glycobiology</i> , 2003, 13, 73-85.	1.3	28
88	Inherited disorders of transition metal metabolism: an update. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 519-529.	1.7	27
89	Rapid, proteomic urine assay for monitoring progressive organ disease in Fabry disease. <i>Journal of Medical Genetics</i> , 2020, 57, 38-47.	1.5	26
90	Structural Determination of Lysosphingomyelin-509 and Discovery of Novel Class Lipids from Patients with Niemannâ€²Pick Disease Type C. <i>International Journal of Molecular Sciences</i> , 2019, 20, 5018.	1.8	25

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91	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.	1.8	24
92	An LC-MS/MS-Based Method for the Quantification of Pyridox(am)ine 5-Phosphate Oxidase Activity in Dried Blood Spots from Patients with Epilepsy. <i>Analytical Chemistry</i> , 2017, 89, 8892-8900.	3.2	24
93	New synthesis of (±)-CMBHC and its confirmation as a metabolite of ±-tocopherol (vitamin E). <i>Bioorganic and Medicinal Chemistry</i> , 2001, 9, 1337-1343.	1.4	23
94	<i>RARS</i> mutations in a sibship with infantile spasms. <i>Epilepsia</i> , 2016, 57, e97-e102.	2.6	23
95	Diversity of congenital disorders of glycosylation. <i>Lancet, The</i> , 2001, 357, 1382-1383.	6.3	22
96	Seizures Due to a <i>KCNQ2</i> Mutation: Treatment with Vitamin B6. <i>JIMD Reports</i> , 2015, 27, 79-84.	0.7	22
97	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
98	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
99	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.	1.7	21
100	Genotype-phenotype correlation in <i>PEX5</i> -deficient peroxisome biogenesis defective cell lines. <i>Human Mutation</i> , 2009, 30, 93-98.	1.1	21
101	Ten novel <i>HMGCL</i> mutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. <i>Human Mutation</i> , 2009, 30, E520-E529.	1.1	21
102	Normal Cerebrospinal Fluid Pyridoxal 5-Phosphate Level in a <i>PNPO</i> -Deficient Patient with Neonatal-Onset Epileptic Encephalopathy. <i>JIMD Reports</i> , 2015, 22, 67-75.	0.7	21
103	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.	3.2	20
104	Organic Solute Transporter Alpha Deficiency: A Disorder With Cholestasis, Liver Fibrosis, and Congenital Diarrhea. <i>Hepatology</i> , 2020, 71, 1879-1882.	3.6	19
105	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
106	Novel Mutations in X-Linked Dominant Chondrodysplasia Punctata ( <i>CDPX2</i> ). <i>Journal of Investigative Dermatology</i> , 2003, 121, 939-942.	0.3	17
107	Mutations in <i>SLC25A22</i> : hyperprolinaemia, vacuolated fibroblasts and presentation with developmental delay. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 385-394.	1.7	16
108	Investigation of diagnostic performance of five urinary cholesterol metabolites for Niemann-Pick disease type C. <i>Journal of Lipid Research</i> , 2019, 60, 2074-2081.	2.0	16

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109	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.	1.5	15
110	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.	1.2	14
111	Deep mining of oxysterols and cholestenic acids in human plasma and cerebrospinal fluid: Quantification using isotope dilution mass spectrometry. <i>Analytica Chimica Acta</i> , 2021, 1154, 338259.	2.6	14
112	Effect of Intravenous Lipid Emulsions on Hepatic Cholesterol Metabolism. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2000, 30, 538-546.	0.9	14
113	Infantile parkinsonism-dystonia: Tyrosine hydroxylase deficiency. <i>Movement Disorders</i> , 1998, 13, 350-350.	2.2	12
114	Diagnostic performance evaluation of sulfate-conjugated cholesterol metabolites as urinary biomarkers of Niemann-Pick disease type C. <i>Clinica Chimica Acta</i> , 2019, 494, 58-63.	0.5	12
115	Bile acid biosynthesis in Smith-Lemli-Opitz syndrome bypassing cholesterol: Potential importance of pathway intermediates. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2021, 206, 105794.	1.2	12
116	Sphincterotomy for Jaundice in a Neonate. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 1996, 23, 507-509.	0.9	11
117	Tubular aggregates caused by serine active site containing 1 ( <i>SERAC1</i> ) mutations in a patient with a mitochondrial encephalopathy. <i>Neuropathology and Applied Neurobiology</i> , 2015, 41, 399-402.	1.8	10
118	Phenotypic variability in a dystonia family with mutations in the manganese transporter gene. <i>Movement Disorders</i> , 2013, 28, 685-686.	2.2	9
119	Coenzyme Q10 and Pyridoxal Phosphate Deficiency Is a Common Feature in Mucopolysaccharidosis Type III. <i>JIMD Reports</i> , 2015, 25, 1-7.	0.7	8
120	Global serum glycoform profiling for the investigation of dystroglycanopathies & Congenital Disorders of Glycosylation. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 7, 55-62.	0.4	8
121	Is susceptibility to severe COVID-19 disease an inborn error of metabolism?. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 906-907.	1.7	8
122	Prenatal testing for a novel EBP missense mutation causing X-linked dominant chondrodysplasia punctata. <i>Prenatal Diagnosis</i> , 2008, 28, 384-388.	1.1	7
123	The effectiveness of correcting abnormal metabolic profiles. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 2-13.	1.7	7
124	Clinical phenotype of desmosterolosis. , 1998, 75, 145.		4
125	Tissue Proteome of 2-Hydroxyacyl-CoA Lyase Deficient Mice Reveals Peroxisome Proliferation and Activation of $\beta$ -Oxidation. <i>International Journal of Molecular Sciences</i> , 2022, 23, 987.	1.8	4
126	Neurotransmitter diseases and related conditions. <i>Molecular Genetics and Metabolism</i> , 2007, 92, 189-197.	0.5	3



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127	A preterm neonate with seizures unresponsive to conventional treatment. <i>BMJ Case Reports</i> , 2015, 2015, bcr2015209743-bcr2015209743.	0.2	3
128	Disorders of Neurotransmission. , 2012, , 405-422.		3
129	Disorders of Bile Acid Synthesis. , 2016, , 465-475.		3
130	Characterization of Novel Pathogenic Variants Causing Pyridox(am)ine 5â€²-Phosphate Oxidase-Dependent Epilepsy. <i>International Journal of Molecular Sciences</i> , 2021, 22, 12013.	1.8	3
131	Concerning â€œAgamanolis diseaseâ€. <i>American Journal of Medical Genetics Part A</i> , 1995, 56, 289-289.	2.4	2
132	â€œAfrican medicineâ€ and Reye's syndrome. <i>Lancet, The</i> , 2004, 363, 860.	6.3	2
133	Disorders of Bile Acid Synthesis. , 2006, , 421-430.		2
134	Correspondence. <i>Pediatric Research</i> , 2003, 53, 865-865.	1.1	1
135	Disorders of Bile Acid Synthesis. , 2006, , 341-351.		1
136	Disorders of Cholesterol Synthesis. , 2012, , 461-471.		1
137	Micronutrients. <i>FIRE Forum for International Research in Education</i> , 2018, 6, 232640981876501.	0.7	0
138	Measurement of Bile Acids as a Marker of the Functionality of iPSC-Derived Hepatocytes. <i>Methods in Molecular Biology</i> , 2019, 1994, 141-147.	0.4	0
139	Disorders of Neurotransmission. , 2006, , 359-372.		0
140	Disorders of Cholesterol Synthesis. , 2006, , 411-420.		0
141	Disorders of Bile Acid Synthesis. , 2012, , 473-484.		0
142	Mass Spectrometry Measurement of Albuminâ€Alpha Fetoprotein Ratio as an Indicator of iPSC-Derived Hepatocyte Differentiation. <i>Methods in Molecular Biology</i> , 2019, 1994, 149-156.	0.4	0