

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 | Mutations in antiquitin in individuals with pyridoxine-dependent seizures. <i>Nature Medicine</i> , 2006, 12, 307-309. | 30.7 | 476 |
| 2 | B6-responsive disorders: A model of vitamin dependency. <i>Journal of Inherited Metabolic Disease</i> , 2006, 29, 317-326. | 3.6 | 364 |
| 3 | 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 |
| 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. | 6.2 | 321 |
| 5 | The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. <i>Lancet Neurology</i> , The, 2011, 10, 721-733. | 10.2 | 290 |
| 6 | Neonatal epileptic encephalopathy caused by mutations in the PNPO gene encoding pyridox(am)ine 5 α -phosphate oxidase. <i>Human Molecular Genetics</i> , 2005, 14, 1077-1086. | 2.9 | 281 |
| 7 | Pyridoxine dependent epilepsy and antiquitin deficiency. <i>Molecular Genetics and Metabolism</i> , 2011, 104, 48-60. | 1.1 | 258 |
| 8 | Hyperinsulinism in short-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency reveals the importance of β -oxidation in insulin secretion. <i>Journal of Clinical Investigation</i> , 2001, 108, 457-465. | 8.2 | 246 |
| 9 | Mutations in the gene encoding peroxisomal β -methylacyl-CoA racemase cause adult-onset sensory motor neuropathy. <i>Nature Genetics</i> , 2000, 24, 188-191. | 21.4 | 241 |
| 10 | Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonism and dystonia. <i>Nature Communications</i> , 2016, 7, 11601. | 12.8 | 233 |
| 11 | Genotypic and phenotypic spectrum of pyridoxine-dependent epilepsy (ALDH7A1 deficiency). <i>Brain</i> , 2010, 133, 2148-2159. | 7.6 | 219 |
| 12 | The Role of Phytosterols in the Pathogenesis of Liver Complications of Pediatric Parenteral Nutrition. <i>Nutrition</i> , 1998, 14, 158-164. | 2.4 | 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. | 6.2 | 206 |
| 14 | Host-Microbe Co-metabolism Dictates Cancer Drug Efficacy in <i>C. elegans</i> . <i>Cell</i> , 2017, 169, 442-456.e18. | 28.9 | 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. | 2.9 | 175 |
| 16 | Manganese and the Brain. <i>International Review of Neurobiology</i> , 2013, 110, 277-312. | 2.0 | 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. | 7.6 | 151 |

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|----|---|-----|-----------|
| 19 | Alström syndrome. <i>Ophthalmology</i> , 1998, 105, 1274-1280. | 5.2 | 150 |
| 20 | Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. <i>Brain</i> , 2014, 137, 44-56. | 7.6 | 143 |
| 21 | Disorders affecting vitamin B ₆ metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 629-646. | 3.6 | 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. | 2.5 | 138 |
| 23 | Disorders of bile acid synthesis. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 593-604. | 3.6 | 134 |
| 24 | Recommendations for the detection and diagnosis of Niemann-Pick disease type C. <i>Neurology: Clinical Practice</i> , 2017, 7, 499-511. | 1.6 | 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. | 6.2 | 118 |
| 26 | Defective galactosylation of serum transferrin in galactosemia. <i>Glycobiology</i> , 1998, 8, 351-357. | 2.5 | 115 |
| 27 | Hepatic cirrhosis, dystonia, polycythaemia and hypermanganesaemia—A new metabolic disorder. <i>Journal of Inherited Metabolic Disease</i> , 2008, 31, 151-163. | 3.6 | 114 |
| 28 | Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. <i>Brain</i> , 2012, 135, 2875-2882. | 7.6 | 114 |
| 29 | Neonatal presentation of coenzyme Q10 deficiency. <i>Journal of Pediatrics</i> , 2001, 139, 456-458. | 1.8 | 112 |
| 30 | 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 |
| 31 | Liver failure associated with mitochondrial DNA depletion. <i>Journal of Hepatology</i> , 1998, 28, 556-563. | 3.7 | 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. | 3.9 | 104 |
| 33 | Pyridoxine responsiveness in novel mutations of the <i>PNPO</i> gene. <i>Neurology</i> , 2014, 82, 1425-1433. | 1.1 | 100 |
| 34 | 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 |
| 35 | Quantitative Charge-Tags for Sterol and Oxysterol Analysis. <i>Clinical Chemistry</i> , 2015, 61, 400-411. | 3.2 | 89 |
| 36 | Clinical and genetic spectrum of pyruvate dehydrogenase deficiency: Dihydrolipoamide acetyltransferase (E2) deficiency. <i>Annals of Neurology</i> , 2005, 58, 234-241. | 5.3 | 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. | 2.1 | 85 |
| 38 | Cholestenolic acids regulate motor neuron survival via liver X receptors. <i>Journal of Clinical Investigation</i> , 2014, 124, 4829-4842. | 8.2 | 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. | 2.8 | 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. | 6.2 | 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. | 3.2 | 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. | 6.2 | 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. | 7.1 | 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. | 2.9 | 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. | 2.7 | 70 |
| 46 | Inborn errors of metabolism causing epilepsy. <i>Developmental Medicine and Child Neurology</i> , 2013, 55, 23-36. | 2.1 | 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. | 1.8 | 68 |
| 48 | Pyridoxal 5'-phosphate in cerebrospinal fluid; factors affecting concentration. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 529-538. | 3.6 | 68 |
| 49 | TRNT1 deficiency: clinical, biochemical and molecular genetic features. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 90. | 2.7 | 64 |
| 50 | Synthesis and analysis of conjugates of the major vitamin E metabolite, α -CEHC. <i>Free Radical Biology and Medicine</i> , 2002, 33, 807-817. | 2.9 | 63 |
| 51 | MAN1B1 Deficiency: An Unexpected CDG-II. <i>PLoS Genetics</i> , 2013, 9, e1003989. | 3.5 | 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 7α -hydroxylase deficiency: successful treatment with chenodeoxycholic acid. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 851-861. | 3.6 | 58 |
| 54 | An intriguing "silent" mutation and a founder effect in <i>antiquitin (ALDH7A1)</i> . <i>Annals of Neurology</i> , 2007, 62, 414-418. | 5.3 | 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. | 2.9 | 56 |
| 56 | Desmosterolosis: a new inborn error of cholesterol biosynthesis. <i>Lancet</i> , The, 1996, 348, 404. | 13.7 | 55 |
| 57 | Optimisation of Bile Production during Normothermic Preservation of Porcine Livers. <i>American Journal of Transplantation</i> , 2002, 2, 593-599. | 4.7 | 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. | 1.1 | 54 |
| 59 | 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 |
| 60 | <i>PDXK</i> mutations cause polyneuropathy responsive to pyridoxal 5-phosphate supplementation. <i>Annals of Neurology</i> , 2019, 86, 225-240. | 5.3 | 54 |
| 61 | Inborn errors presenting with liver dysfunction. <i>Seminars in Fetal and Neonatal Medicine</i> , 2002, 7, 49-63. | 2.7 | 53 |
| 62 | Identification of \pm 1-Antitrypsin Variants in Plasma with the Use of Proteomic Technology. <i>Clinical Chemistry</i> , 2001, 47, 2012-2022. | 3.2 | 52 |
| 63 | Analysis of mutant DNA polymerase β in patients with mitochondrial DNA depletion. <i>Human Mutation</i> , 2009, 30, 248-254. | 2.5 | 52 |
| 64 | Vps33b is crucial for structural and functional hepatocyte polarity. <i>Journal of Hepatology</i> , 2017, 66, 1001-1011. | 3.7 | 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. | 2.3 | 51 |
| 66 | Variable Clinical Spectrum of the Most Common Inborn Error of Bile Acid Metabolism: 3-hydroxy- Δ^5 - Δ^27 -steroid Dehydrogenase Deficiency. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2010, 50, 61-66. | 1.8 | 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. | 2.9 | 50 |
| 68 | Human Δ^4 -3-oxosteroid 5-reductase (AKR1D1) deficiency and steroid metabolism. <i>Steroids</i> , 2008, 73, 417-423. | 1.8 | 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. | 3.6 | 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. | 3.6 | 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. | 2.2 | 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. | 2.2 | 44 |

| # | ARTICLE | IF | CITATIONS |
|----|---|-----|-----------|
| 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. | 1.1 | 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. | 3.6 | 42 |
| 75 | First Trimester Prenatal Diagnosis of Smith-Lemli-Opitz Syndrome(7-Dehydrocholesterol Reductase) Tj ETQq1 1 0.784314 rgBT/Overl 2.3 42 | 2.3 | 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. | 1.5 | 40 |
| 77 | Differential diagnosis in patients with suspected bile acid synthesis defects. <i>World Journal of Gastroenterology</i> , 2012, 18, 1067. | 3.3 | 38 |
| 78 | Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. <i>Brain</i> , 2016, 139, 2844-2854. | 7.6 | 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. | 3.8 | 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. | 1.1 | 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. | 2.5 | 34 |
| 82 | Diagnosis of congenital disorders of glycosylation type-I using protein chip technology. <i>Proteomics</i> , 2006, 6, 2295-2304. | 2.2 | 33 |
| 83 | Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. <i>Neurology</i> , 2014, 83, 1873-1875. | 1.1 | 33 |
| 84 | Pyridoxal 5â€²-phosphate deficiency causes a loss of aromatic l-amino acid decarboxylase in patients and human neuroblastoma cells, implications for aromatic l-amino acid decarboxylase and vitamin B ₆ deficiency states. <i>Journal of Neurochemistry</i> , 2010, 114, 87-96. | 3.9 | 31 |
| 85 | Transaldolase deficiency in a two-year-old boy with cirrhosis. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 255-258. | 1.1 | 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. | 2.7 | 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. | 2.5 | 28 |
| 88 | Inherited disorders of transition metal metabolism: an update. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 519-529. | 3.6 | 27 |
| 89 | Rapid, proteomic urine assay for monitoring progressive organ disease in Fabry disease. <i>Journal of Medical Genetics</i> , 2020, 57, 38-47. | 3.2 | 26 |
| 90 | Structural Determination of Lysosphingomyelin-509 and Discovery of Novel Class Lipids from Patients with Niemannâ€”Pick Disease Type C. <i>International Journal of Molecular Sciences</i> , 2019, 20, 5018. | 4.1 | 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. | 3.7 | 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. | 6.5 | 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. | 3.0 | 23 |
| 94 | <i>RARS</i> mutations in a sibship with infantile spasms. <i>Epilepsia</i> , 2016, 57, e97-e102. | 5.1 | 23 |
| 95 | Diversity of congenital disorders of glycosylation. <i>Lancet, The</i> , 2001, 357, 1382-1383. | 13.7 | 22 |
| 96 | Seizures Due to a <i>KCNQ2</i> Mutation: Treatment with Vitamin B6. <i>JIMD Reports</i> , 2015, 27, 79-84. | 1.5 | 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. | 3.7 | 21 |
| 100 | Genotype-phenotype correlation in <i>PEX5</i> -deficient peroxisome biogenesis defective cell lines. <i>Human Mutation</i> , 2009, 30, 93-98. | 2.5 | 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. | 2.5 | 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. | 1.5 | 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. | 6.5 | 20 |
| 104 | Organic Solute Transporter Alpha Deficiency: A Disorder With Cholestasis, Liver Fibrosis, and Congenital Diarrhea. <i>Hepatology</i> , 2020, 71, 1879-1882. | 7.3 | 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. | 0.7 | 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. | 3.6 | 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. | 4.2 | 16 |

| # | ARTICLE | IF | CITATIONS |
|-----|--|-----|-----------|
| 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. | 1.2 | 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. | 2.4 | 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. | 5.4 | 14 |
| 112 | Effect of Intravenous Lipid Emulsions on Hepatic Cholesterol Metabolism. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2000, 30, 538-546. | 1.8 | 14 |
| 113 | Infantile parkinsonism-dystonia: Tyrosine hydroxylase deficiency. <i>Movement Disorders</i> , 1998, 13, 350-350. | 3.9 | 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. | 1.1 | 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. | 2.5 | 12 |
| 116 | Sphincterotomy for Jaundice in a Neonate. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 1996, 23, 507-509. | 1.8 | 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. | 3.2 | 10 |
| 118 | Phenotypic variability in a dystonia family with mutations in the manganese transporter gene. <i>Movement Disorders</i> , 2013, 28, 685-686. | 3.9 | 9 |
| 119 | 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 |
| 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. | 1.1 | 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. | 3.6 | 8 |
| 122 | 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 |
| 123 | The effectiveness of correcting abnormal metabolic profiles. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 2-13. | 3.6 | 7 |
| 124 | Clinical phenotype of desmosterolosis. <i>American Journal of Medical Genetics Part A</i> , 1998, 75, 145-152. | 2.4 | 4 |
| 125 | Tissue Proteome of 2-Hydroxyacyl-CoA Lyase Deficient Mice Reveals Peroxisome Proliferation and Activation of β -Oxidation. <i>International Journal of Molecular Sciences</i> , 2022, 23, 987. | 4.1 | 4 |
| 126 | Neurotransmitter diseases and related conditions. <i>Molecular Genetics and Metabolism</i> , 2007, 92, 189-197. | 1.1 | 3 |

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|-----|---|------|-----------|
| 127 | A preterm neonate with seizures unresponsive to conventional treatment. <i>BMJ Case Reports</i> , 2015, 2015, bcr2015209743-bcr2015209743. | 0.5 | 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. | 4.1 | 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. | 13.7 | 2 |
| 133 | Disorders of Bile Acid Synthesis. , 2006, , 421-430. | | 2 |
| 134 | Correspondence. <i>Pediatric Research</i> , 2003, 53, 865-865. | 2.3 | 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. | 0.9 | 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. | 0.9 | 0 |