FrÃ©dÃ©ric M Vaz

List of Publications by Year
in descending order

165 papers	8,934 citations	47004 47 h-index	48312 88 g-index
174 all docs	174 docs citations	174 times ranked	11550 citing authors

#	Article	IF	Citations
1	Modeling the mitochondrial cardiomyopathy of Barth syndrome with induced pluripotent stem cell and heart-on-chip technologies. Nature Medicine, 2014, 20, 616-623.	30.7	733
2	Acylcarnitines. Diabetes, 2013, 62, 1-8.	0.6	551
3	Carnitine biosynthesis in mammals. Biochemical Journal, 2002, 361, 417-429.	3.7	527
4	Carnitine biosynthesis in mammals. Biochemical Journal, 2002, 361, 417.	3.7	372
5	Cardiolipin provides an essential activating platform for caspase-8 on mitochondria. Journal of Cell Biology, 2008, 183, 681-696.	5.2	258
6	Xâ€linked cardioskeletal myopathy and neutropenia (Barth syndrome): An update. American Journal of Medical Genetics Part A, 2004, 126A, 349-354.	2.4	244
7	Cardiac and Skeletal Muscle Defects in a Mouse Model of Human Barth Syndrome. Journal of Biological Chemistry, 2011, 286, 899-908.	3.4	226
8	Mutations in LPIN1 Cause Recurrent Acute Myoglobinuria in Childhood. American Journal of Human Genetics, 2008, 83, 489-494.	6.2	189
9	Aberrant cardiolipin metabolism in the yeast taz1 mutant: a model for Barth syndrome. Molecular Microbiology, 2003, 51, 149-158.	2.5	186
10	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nature Genetics, 2012, 44, 797-802.	21.4	175
11	Sodium taurocholate cotransporting polypeptide (SLC10A1) deficiency: Conjugated hypercholanemia without a clear clinical phenotype. Hepatology, 2015, 61, 260-267.	7.3	169
12	Barth syndrome: Cellular compensation of mitochondrial dysfunction and apoptosis inhibition due to changes in cardiolipin remodeling linked to tafazzin (TAZ) gene mutation. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 1194-1206.	3.8	140
13	Cardiolipin deficiency affects respiratory chain function and organization in an induced pluripotent stem cell model of Barth syndrome. Stem Cell Research, 2013, 11, 806-819.	0.7	140
14	The enigmatic role of tafazzin in cardiolipin metabolism. Biochimica Et Biophysica Acta - Biomembranes, 2009, 1788, 2003-2014.	2.6	135
15	Monolysocardiolipins accumulate in Barth syndrome but do not lead to enhanced apoptosis. Journal of Lipid Research, 2005, 46, 1182-1195.	4.2	124
16	Cardiolipin and monolysocardiolipin analysis in fibroblasts, lymphocytes, and tissues using high-performance liquid chromatographyâ€“mass spectrometry as a diagnostic test for Barth syndrome. Analytical Biochemistry, 2009, 387, 230-237.	2.4	120
17	Only One Splice Variant of the Human TAZ Gene Encodes a Functional Protein with a Role in Cardiolipin Metabolism. Journal of Biological Chemistry, 2003, 278, 43089-43094.	3.4	118
18	A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 7974-7981.	7.1	118

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19	LPIN1 gene mutations: a major cause of severe rhabdomyolysis in early childhood. Human Mutation, 2010, 31, E1564-E1573.	2.5	112
20	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. American Journal of Human Genetics, 2015, 96, 245-257.	6.2	111
21	Bloodspot Assay Using HPLCâ€“Tandem Mass Spectrometry for Detection of Barth Syndrome. Clinical Chemistry, 2008, 54, 371-378.	3.2	108
22	Bile acid analysis in human disorders of bile acid biosynthesis. Molecular Aspects of Medicine, 2017, 56, 10-24.	6.4	102
23	The Human TAZ Gene Complements Mitochondrial Dysfunction in the Yeast taz1Î” Mutant. Journal of Biological Chemistry, 2004, 279, 44394-44399.	3.4	96
24	Lipoprotein Lipase Maintains Microglial Innate Immunity in Obesity. Cell Reports, 2017, 20, 3034-3042.	6.4	89
25	Identification and characterization of human cardiolipin synthase. FEBS Letters, 2006, 580, 3059-3064.	2.8	87
26	Enzymology of the carnitine biosynthesis pathway. IUBMB Life, 2010, 62, 357-362.	3.4	87
27	PPARÎ±-activation results in enhanced carnitine biosynthesis and OCTN2-mediated hepatic carnitine accumulation. Biochimica Et Biophysica Acta - Bioenergetics, 2007, 1767, 1134-1142.	1.0	86
28	Retrospective study of long-term outcomes of enzyme replacement therapy in Fabry disease: Analysis of prognostic factors. PLoS ONE, 2017, 12, e0182379.	2.5	83
29	Peroxisomes can oxidize mediumâ€•and longâ€•chain fatty acids through a pathway involving ABCD3 and HSD17B4. FASEB Journal, 2019, 33, 4355-4364.	0.5	82
30	Characterization of carnitine and fatty acid metabolism in the long-chain acyl-CoA dehydrogenase-deficient mouse. Biochemical Journal, 2005, 387, 185-193.	3.7	81
31	Growth and Fatty Acid Profiles of VLBW Infants Receiving a Multicomponent Lipid Emulsion From Birth. Journal of Pediatric Gastroenterology and Nutrition, 2014, 58, 417-427.	1.8	76
32	Mutations in PCYT2 disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia. Brain, 2019, 142, 3382-3397.	7.6	76
33	Zellweger spectrum disorders: clinical manifestations in patients surviving into adulthood. Journal of Inherited Metabolic Disease, 2016, 39, 93-106.	3.6	73
34	Agalsidase alfa versus agalsidase beta for the treatment of Fabry disease: an international cohort study. Journal of Medical Genetics, 2018, 55, 351-358.	3.2	72
35	Distinct effects of tafazzin deletion in differentiated and undifferentiated mitochondria. Mitochondrion, 2009, 9, 86-95.	3.4	68
36	Clinical and biochemical characterization of four patients with mutations in ECHS1. Orphanet Journal of Rare Diseases, 2015, 10, 79.	2.7	68

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37	Molecular and Biochemical Characterization of Rat Î³-Trimethylaminobutyraldehyde Dehydrogenase and Evidence for the Involvement of Human Aldehyde Dehydrogenase 9 in Carnitine Biosynthesis. Journal of Biological Chemistry, 2000, 275, 7390-7394.	3.4	65
38	Lipidomic analysis of fibroblasts from Zellweger spectrum disorder patients identifies disease-specific phospholipid ratios. Journal of Lipid Research, 2016, 57, 1447-1454.	4.2	65
39	Favourable effect of early versus late start of enzyme replacement therapy on plasma globotriaosylsphingosine levels in men with classical Fabry disease. Molecular Genetics and Metabolism, 2017, 121, 157-161.	1.1	64
40	A sensitive mass spectrometry platform identifies metabolic changes of life history traits in C. elegans. Scientific Reports, 2017, 7, 2408.	3.3	61
41	Bactericidal activity of amphipathic cationic antimicrobial peptides involves altering the membrane fluidity when interacting with the phospholipid bilayer. Biochimica Et Biophysica Acta - Biomembranes, 2018, 1860, 2404-2415.	2.6	59
42	Catecholamines profiles at diagnosis: Increased diagnostic sensitivity and correlation with biological and clinical features in neuroblastoma patients. European Journal of Cancer, 2017, 72, 235-243.	2.8	57
43	The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome. Orphanet Journal of Rare Diseases, 2017, 12, 49.	2.7	57
44	Deletion of the Cardiolipin-specific Phospholipase Cld1 Rescues Growth and Life Span Defects in the Tafazzin Mutant. Journal of Biological Chemistry, 2014, 289, 3114-3125.	3.4	55
45	Defining functional classes of Barth syndrome mutation in humans. Human Molecular Genetics, 2016, 25, 1754-1770.	2.9	53
46	Barth syndrome cells display widespread remodeling of mitochondrial complexes without affecting metabolic flux distribution. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3650-3658.	3.8	53
47	Pyruvate dehydrogenase complex plays a central role in brown adipocyte energy expenditure and fuel utilization during short-term beta-adrenergic activation. Scientific Reports, 2018, 8, 9562.	3.3	53
48	Comparison of C26:0-carnitine and C26:0-lysophosphatidylcholine as diagnostic markers in dried blood spots from newborns and patients with adrenoleukodystrophy. Molecular Genetics and Metabolism, 2017, 122, 209-215.	1.1	50
49	Carnitine Biosynthesis: Identification of the cDNA Encoding Human Î³-Butyrobetaine Hydroxylase. Biochemical and Biophysical Research Communications, 1998, 250, 506-510.	2.1	49
50	Functional characterisation of peroxisomal Î²â€oxidation disorders in fibroblasts using lipidomics. Journal of Inherited Metabolic Disease, 2018, 41, 479-487.	3.6	48
51	Adrenoleukodystrophy Newborn Screening in the Netherlands (SCAN Study): The X-Factor. Frontiers in Cell and Developmental Biology, 2020, 8, 499.	3.7	47
52	Molecular and Biochemical Characterization of Rat Îµ-N-Trimethyllysine Hydroxylase, the First Enzyme of Carnitine Biosynthesis. Journal of Biological Chemistry, 2001, 276, 33512-33517.	3.4	46
53	The important role of biochemical and functional studies in the diagnostics of peroxisomal disorders. Journal of Inherited Metabolic Disease, 2016, 39, 531-543.	3.6	44
54	Delineating the role of alterations in lipid metabolism to the pathogenesis of inherited skeletal and cardiac muscle disorders. Journal of Lipid Research, 2012, 53, 4-27.	4.2	43

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55	Long-Term Dose-Dependent Agalsidase Effects on Kidney Histology in Fabry Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2017, 12, 1470-1479.	4.5	42
56	Organic solute transporterâ€Î² (SLC51B) deficiency in two brothers with congenital diarrhea and features of cholestasis. Hepatology, 2018, 68, 590-598.	7.3	41
57	Tissue Carnitine Homeostasis in Very-Long-Chain Acyl-CoA Dehydrogenaseâ€"Deficient Mice. Pediatric Research, 2005, 57, 760-764.	2.3	39
58	Fatty acid metabolism and its longitudinal relationship with the hypothalamicâ€"pituitaryâ€"adrenal axis in major depression: Associations with prospective antidepressant response. Psychoneuroendocrinology, 2015, 59, 1-13.	2.7	37
59	Cholic acid therapy in Zellweger spectrum disorders. Journal of Inherited Metabolic Disease, 2016, 39, 859-868.	3.6	37
60	Evaluation of C26:0â€lysophosphatidylcholine and C26:0â€carnitine as diagnostic markers for Zellweger spectrum disorders. Journal of Inherited Metabolic Disease, 2017, 40, 875-881.	3.6	37
61	Principles and practice of lipidomics. Journal of Inherited Metabolic Disease, 2015, 38, 41-52.	3.6	36
62	New clinical and molecular insights on Barth syndrome. Orphanet Journal of Rare Diseases, 2013, 8, 27.	2.7	35
63	Mitochondrial Fatty Acid Oxidation Disorders: Laboratory Diagnosis, Pathogenesis, and the Complicated Route to Treatment. Journal of Lipid and Atherosclerosis, 2020, 9, 313.	3.5	35
64	C26:0-Carnitine Is a New Biomarker for X-Linked Adrenoleukodystrophy in Mice and Man. PLoS ONE, 2016, 11, e0154597.	2.5	33
65	Rapid screening for lipid storage disorders using biochemical markers. Expert center data and review of the literature. Molecular Genetics and Metabolism, 2018, 123, 76-84.	1.1	33
66	Measurement of carnitine biosynthesis enzyme activities by tandem mass spectrometry: Differences between the mouse and the rat. Analytical Biochemistry, 2006, 354, 132-139.	2.4	32
67	Identification and characterization of a complete carnitine biosynthesis pathway inCandida albicans. FASEB Journal, 2009, 23, 2349-2359.	0.5	32
68	New targets for monitoring and therapy in Barth syndrome. Genetics in Medicine, 2016, 18, 1001-1010.	2.4	32
69	Comparison of the Diagnostic Performance of C26:0-Lysophosphatidylcholine and Very Long-Chain Fatty Acids Analysis for Peroxisomal Disorders. Frontiers in Cell and Developmental Biology, 2020, 8, 690.	3.7	31
70	Skeletal muscle in healthy humans exhibits a day-night rhythm in lipid metabolism. Molecular Metabolism, 2020, 37, 100989.	6.5	30
71	Vulnerability for new episodes in recurrent major depressive disorder: protocol for the longitudinal DELTA-neuroimaging cohort study. BMJ Open, 2016, 6, e009510.	1.9	29
72	The cellular and molecular mechanisms for neutropenia in Barth syndrome. European Journal of Haematology, 2012, 88, 195-209.	2.2	28

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73	Levels of Red Blood Cell Fatty Acids in Patients With Psychosis, Their Unaffected Siblings, and Healthy Controls. Schizophrenia Bulletin, 2016, 42, 358-368.	4.3	28
74	A newborn screening method for cerebrotendinous xanthomatosis using bile alcohol glucuronides and metabolite ratios. Journal of Lipid Research, 2017, 58, 1002-1007.	4.2	28
75	SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family. Journal of Medical Genetics, 2018, 55, 39-47.	3.2	28
76	NTCP deficiency and persistently raised bile salts: an adult case. Journal of Inherited Metabolic Disease, 2017, 40, 313-315.	3.6	27
77	Metabolomics and lipidomics in Caenorhabditis elegans using a single-sample preparation. DMM Disease Models and Mechanisms, 2021, 14, .	2.4	27
78	Hepatotoxicity due to chenodeoxycholic acid supplementation in an infant with cerebrotendinous xanthomatosis: implications for treatment. European Journal of Pediatrics, 2016, 175, 143-146.	2.7	26
79	Metabolic differences between bronchial epithelium from healthy individuals and patients with asthma and the effect of bronchial thermoplasty. Journal of Allergy and Clinical Immunology, 2021, 148, 1236-1248.	2.9	26
80	Fatty Acid Oxidation in Peroxisomes: Enzymology, Metabolic Crosstalk with Other Organelles and Peroxisomal Disorders. Advances in Experimental Medicine and Biology, 2020, 1299, 55-70.	1.6	26
81	Mitochondrial disruption in peroxisome deficient cells is hepatocyte selective but is not mediated by common hepatic peroxisomal metabolites. Mitochondrion, 2018, 39, 51-59.	3.4	26
82	Lipidomics unveils lipid dyshomeostasis and low circulating plasmalogens as biomarkers in a monogenic mitochondrial disorder. JCI Insight, 2019, 4, .	5.0	26
83	Integrative Analysis of the Inflammatory Bowel Disease Serum Metabolome Improves Our Understanding of Genetic Etiology and Points to Novel Putative Therapeutic Targets. Gastroenterology, 2022, 162, 828-843.e11.	1.3	26
84	Indoleamine 2,3â€dioxygenase (<scp>IDO</scp>)â€1 and <scp>IDO</scp>â€2 activity and severe course of <scp>COVID</scp>â€19. Journal of Pathology, 2022, 256, 256-261.	4.5	26
85	Toward newborn screening of cerebrotendinous xanthomatosis: results of a biomarker research study using 32,000 newborn dried blood spots. Genetics in Medicine, 2020, 22, 1606-1612.	2.4	25
86	Disturbed brain ether lipid metabolism and histology in <scp>SjÃ¶grenâ€Larsson</scp> syndrome. Journal of Inherited Metabolic Disease, 2020, 43, 1265-1278.	3.6	25
87	The effect of mirabegron on energy expenditure and brown adipose tissue in healthy lean South <scp>Asian and Europid</scp> men. Diabetes, Obesity and Metabolism, 2020, 22, 2032-2044.	4.4	25
88	An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. Genetics in Medicine, 2021, 23, 740-750.	2.4	25
89	The dynamics of cardiolipin synthesis post-mitochondrial fusion. Biochimica Et Biophysica Acta - Biomembranes, 2010, 1798, 1577-1585.	2.6	23
90	Mitochondrial Encephalopathy and Transient 3-Methylglutaconic Aciduria in ECHS1 Deficiency: Long-Term Follow-Up. JIMD Reports, 2017, 39, 83-87.	1.5	23

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91	Ethnic differences in metabolite signatures and type 2 diabetes: a nested caseâ€“control analysis among people of South Asian, African and European origin. Nutrition and Diabetes, 2017, 7, 300.	3.2	23
92	A mutation creating an upstream translation initiation codon in SLC22A5 5â€²UTR is a frequent cause of primary carnitine deficiency. Human Mutation, 2019, 40, 1899-1904.	2.5	23
93	Plasma lipidomics as a diagnostic tool for peroxisomal disorders. Journal of Inherited Metabolic Disease, 2018, 41, 489-498.	3.6	22
94	Lipidomics in Nonalcoholic Fatty Liver Disease. Journal of Pediatric Gastroenterology and Nutrition, 2020, 71, 433-439.	1.8	22
95	Carnitine biosynthesis inNeurospora crassa: identification of a cDNA coding for Ã‰Â›-N-trimethyllysine hydroxylase and its functional expression inSaccharomyces cerevisiae. FEMS Microbiology Letters, 2002, 210, 19-23.	1.8	21
96	Cardiolipin Molecular Species with Shorter Acyl Chains Accumulate in Saccharomyces cerevisiae Mutants Lacking the Acyl Coenzyme A-binding Protein Acb1p. Journal of Biological Chemistry, 2009, 284, 27609-27619.	3.4	21
97	Translational Metabolism: A multidisciplinary approach towards precision diagnosis of inborn errors of metabolism in the omics era. Journal of Inherited Metabolic Disease, 2019, 42, 197-208.	3.6	20
98	An improved enzyme assay for carnitine palmitoyl transferase I in fibroblasts using tandem mass spectrometry. Molecular Genetics and Metabolism, 2007, 90, 24-29.	1.1	19
99	Urinary metabolic phenotyping of mucopolysaccharidosis type I combining untargeted and targeted strategies with data modeling. Clinica Chimica Acta, 2017, 475, 7-14.	1.1	19
100	Submitochondrial localization of 6â€‹N€trimethyllysine dioxygenaseâ€ƒâ€”implications for carnitine biosynthesis. FEBS Journal, 2007, 274, 5845-5851.	4.7	18
101	Update on newborn dried bloodspot testing for cerebrotendinous xanthomatosis: An available high-throughput liquid-chromatography tandem mass spectrometry method. Molecular Genetics and Metabolism Reports, 2016, 7, 11-15.	1.1	18
102	The cholic acid extension study in Zellweger spectrum disorders: Results and implications for therapy. Journal of Inherited Metabolic Disease, 2019, 42, 303-312.	3.6	18
103	Cerebellar and hepatic alterations in ACBD5-deficient mice are associated with unexpected, distinct alterations in cellular lipid homeostasis. Communications Biology, 2020, 3, 713.	4.4	18
104	Cardiolipin Remodeling Defects Impair Mitochondrial Architecture and Function in a Murine Model of Barth Syndrome Cardiomyopathy. Circulation: Heart Failure, 2021, 14, e008289.	3.9	17
105	Contributions of amino acid, acylcarnitine and sphingolipid profiles to type 2 diabetes risk among South-Asian Surinamese and Dutch adults. BMJ Open Diabetes Research and Care, 2020, 8, e001003.	2.8	16
106	Plasma oxalate: comparison of methodologies. Urolithiasis, 2020, 48, 473-480.	2.0	16
107	Aging selectively dampens oscillation of lipid abundance in white and brown adipose tissue. Scientific Reports, 2021, 11, 5932.	3.3	16
108	Mouse Tafazzin Is Required for Male Germ Cell Meiosis and Spermatogenesis. PLoS ONE, 2015, 10, e0131066.	2.5	15

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109	The impact of altered carnitine availability on acylcarnitine metabolism, energy expenditure and glucose tolerance in diet-induced obese mice. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 1375-1382.	3.8	15
110	Biochemical and imaging parameters in acid sphingomyelinase deficiency: Potential utility as biomarkers. Molecular Genetics and Metabolism, 2020, 130, 16-26.	1.1	15
111	Analysis of carnitine biosynthesis metabolites in urine by HPLC-electrospray tandem mass spectrometry. Clinical Chemistry, 2002, 48, 826-34.	3.2	15
112	Polar metabolomics in human muscle biopsies using a liquid-liquid extraction and full-scan LC-MS. STAR Protocols, 2022, 3, 101302.	1.2	15
113	Polyunsaturated fatty acid biostatus, phospholipase A2 activity and brain white matter microstructure across adolescence. Neuroscience, 2017, 343, 423-433.	2.3	14
114	Multi-Omic Approach to Identify Phenotypic Modifiers Underlying Cerebral Demyelination in X-Linked Adrenoleukodystrophy. Frontiers in Cell and Developmental Biology, 2020, 8, 520.	3.7	14
115	Functional analysis of TMLH variants and definition of domains required for catalytic activity and mitochondrial targeting. Journal of Cellular Physiology, 2005, 204, 839-847.	4.1	13
116	Slc22a5 haploinsufficiency does not aggravate the phenotype of the longâ€chain acylâ€CoA dehydrogenase KO mouse. Journal of Inherited Metabolic Disease, 2020, 43, 486-495.	3.6	13
117	Identification of Metabolic Biomarkers in Relation to Methotrexate Response in Early Rheumatoid Arthritis. Journal of Personalized Medicine, 2020, 10, 271.	2.5	13
118	ECHS1 disease in two unrelated families of Samoan descent: Common variant â€rare disorder. American Journal of Medical Genetics, Part A, 2021, 185, 157-167.	1.2	13
119	Inherited disorders of complex lipid metabolism: A clinical review. Journal of Inherited Metabolic Disease, 2021, 44, 809-825.	3.6	13
120	Cell Type-Selective Loss of Peroxisomal Î²-Oxidation Impairs Bipolar Cell but Not Photoreceptor Survival in the Retina. Cells, 2022, 11, 161.	4.1	13
121	The Platelet Lipidome Is Altered in Patients with COVID-19 and Correlates with Platelet Reactivity. Thrombosis and Haemostasis, 2022, 122, 1683-1692.	3.4	13
122	Supplementation with a powdered blend of PUFAs normalizes DHA and AA levels in patients with PKU. Molecular Genetics and Metabolism, 2013, 109, 121-124.	1.1	12
123	Oral therapy for riboflavin transporter deficiency - What is the regimen of choice?. Parkinsonism and Related Disorders, 2019, 61, 245-247.	2.2	12
124	Peroxisomal Multifunctional Protein 2 Deficiency Perturbs Lipid Homeostasis in the Retina and Causes Visual Dysfunction in Mice. Frontiers in Cell and Developmental Biology, 2021, 9, 632930.	3.7	12
125	PTPMT1 Is Required for Embryonic Cardiac Cardiolipin Biosynthesis to Regulate Mitochondrial Morphogenesis and Heart Development. Circulation, 2021, 144, 403-406.	1.6	12
126	An improved functional assay in blood spot to diagnose Barth syndrome using the monolysocardiolipin/cardiolipin ratio. Journal of Inherited Metabolic Disease, 2022, 45, 29-37.	3.6	11

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127	Bile Acid Alters Male Mouse Fertility in Metabolic Syndrome Context. PLoS ONE, 2015, 10, e0139946.	2.5	11
128	Timeâ€restricted feeding during the inactive phase abolishes the daily rhythm in mitochondrial respiration in rat skeletal muscle. FASEB Journal, 2022, 36, e22133.	0.5	11
129	Deleterious variants in CRLS1 lead to cardiolipin deficiency and cause an autosomal recessive multi-system mitochondrial disease. Human Molecular Genetics, 2022, 31, 3597-3612.	2.9	11
130	Carnitine Biosynthesis. Advances in Experimental Medicine and Biology, 2002, , 117-124.	1.6	10
131	Reliable Diagnosis of Carnitine Palmitoyltransferase Type IA Deficiency by Analysis of Plasma Acylcarnitine Profiles. JIMD Reports, 2016, 32, 33-39.	1.5	10
132	Enantiomerâ€specific pharmacokinetics of D,Lâ€3â€hydroxybutyrate: Implications for the treatment of multiple <scp>acylâ€CoA</scp> dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 926-938.	3.6	10
133	Biochemical Screening of Intellectually Disabled Patients: A Stepping Stone to Initiate a Newborn Screening Program in Pakistan. Frontiers in Neurology, 2019, 10, 762.	2.4	9
134	Differential effects of a 40-hour fast and bile acid supplementation on human GLP-1 and FGF19 responses. American Journal of Physiology - Endocrinology and Metabolism, 2019, 317, E494-E502.	3.5	9
135	Mild Zellweger syndrome due to functionally confirmed novel PEX1 variants. Journal of Applied Genetics, 2020, 61, 87-91.	1.9	9
136	ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. American Journal of Human Genetics, 2021, 108, 2017-2023.	6.2	9
137	Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation?. JIMD Reports, 2015, 29, 89-93.	1.5	8
138	Extended Abstract: Deficiency of Sodium Taurocholate Cotransporting Polypeptide (SLC10A1): A New Inborn Error of Metabolism with an Attenuated Phenotype. Digestive Diseases, 2017, 35, 259-260.	1.9	8
139	Exploring the metabolic fate of medium-chain triglycerides in healthy individuals using a stable isotope tracer. Clinical Nutrition, 2021, 40, 1396-1404.	5.0	8
140	Circadian misalignment disturbs the skeletal muscle lipidome in healthy young men. FASEB Journal, 2021, 35, e21611.	0.5	8
141	Dietary fat and fiber interactively modulate apoptosis and mitochondrial bioenergetic profiles in mouse colon in a site-specific manner. European Journal of Cancer Prevention, 2017, 26, 301-308.	1.3	7
142	Diverse mitochondrial abnormalities in a new cellular model of TAFFAZZIN deficiency are remediated by cardiolipin-interacting small molecules. Journal of Biological Chemistry, 2021, 297, 101005.	3.4	7
143	Mice with a deficiency in Peroxisomal Membrane Protein 4 (PXMP4) display mild changes in hepatic lipid metabolism. Scientific Reports, 2022, 12, 2512.	3.3	7
144	Biochemical Studies in Fibroblasts to Interpret Variants of Unknown Significance in the ABCD1 Gene. Genes, 2021, 12, 1930.	2.4	6

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145	Identification and diagnostic value of phytanoyl- and pristanoyl-carnitine in plasma from patients with peroxisomal disorders. Molecular Genetics and Metabolism, 2017, 121, 279-282.	1.1	5
146	Recommendations for newborn screening for galactokinase deficiency: A systematic review and evaluation of Dutch newborn screening data. Molecular Genetics and Metabolism, 2018, 124, 50-56.	1.1	5
147	Monitoring phenylalanine concentrations in the followâ€up of phenylketonuria patients: An inventory of preâ€analytical and analytical variation. JIMD Reports, 2021, 58, 70-79.	1.5	5
148	Intra-individual plasticity of the TAZ gene leading to different heritable mutations in siblings with Barth syndrome. European Journal of Human Genetics, 2015, 23, 1708-1712.	2.8	4
149	The Association of Acylcarnitines and Amino Acids With Age in Dutch and South-Asian Surinamese Living in Amsterdam. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 3783-3791.	3.6	4
150	Meijer and Vloedmanâ€™s histochemical demonstration of mitochondrial coupling obeys Lambertâ€“Beerâ€™s law in the myocardium. Histochemistry and Cell Biology, 2019, 151, 85-90.	1.7	4
151	Delayed appearance of 3â€methylglutaconic aciduria in neonates with early onset metabolic cardiomyopathies: A potential pitfall for the diagnosis. American Journal of Medical Genetics, Part A, 2020, 182, 64-70.	1.2	4
152	Reduced ech-6 expression attenuates fat-induced lifespan shortening in C. elegans. Scientific Reports, 2022, 12, 3350.	3.3	4
153	Identification of three novel pathogenic mutations in cystathionine beta-synthase gene of Pakistani intellectually disabled patients. Journal of Pediatric Endocrinology and Metabolism, 2021, .	0.9	4
154	Laboratory Diagnosis of Peroxisomal Disorders in the -Omics Era and the Continued Importance of Biomarkers and Biochemical Studies. FIRE Forum for International Research in Education, 2018, 6, 232640981881028.	0.7	3
155	Biallelic variants in TAMM41 are associated with low muscle cardiolipin levels, leading to neonatal mitochondrial disease. Human Genetics and Genomics Advances, 2022, 3, 100097.	1.7	3
156	The cholic acid extension study in Zellweger spectrum disorders: results and implications for therapy. Journal of Inherited Metabolic Disease, 2018, , .	3.6	2
157	Neonatal carnitine concentrations in relation to gestational age and weight. JIMD Reports, 2020, 56, 95-104.	1.5	2
158	Dioxygenases of Carnitine Biosynthesis: 6-N-Trimethyllysine and Î³-Butyrobetaine Hydroxylases. 2-Oxoglutarate-Dependent Oxygenases, 2015, , 324-337.	0.8	2
159	Mutations in LPIN1 Cause Recurrent Acute Myoglobinuria in Childhood. American Journal of Human Genetics, 2009, 84, 95.	6.2	1
160	Reply: Expanding the clinical and genetic spectrum of PCYT2-related disorders. Brain, 2020, 143, e77-e77.	7.6	1
161	Barth syndrome and the many fascinating aspects of cardiolipin. Journal of Inherited Metabolic Disease, 2022, 45, 1-2.	3.6	1
162	Adaptations of the 3T3-L1 adipocyte lipidome to defective ether lipid catabolism uponÂAgmoÂknockdown. Journal of Lipid Research, 2022, 63, 100222.	4.2	1

#	Article	IF	Citations
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