Frdric M Vaz

List of Publications by Citations

162 papers	6,599 citations	40 h-index	78 g-index
174 ext. papers	7,946 ext. citations	5.2 avg, IF	5.66 L-index

#	Paper	IF	Citations
162	Modeling the mitochondrial cardiomyopathy of Barth syndrome with induced pluripotent stem cell and heart-on-chip technologies. *Nature Medicine*, **2014**, 20, 616-23	50.5	604
161	Carnitine biosynthesis in mammals. *Biochemical Journal*, **2002**, 361, 417-429	3.8	464
160	Acylcarnitines: reflecting or inflicting insulin resistance?. *Diabetes*, **2013**, 62, 1-8	0.9	385
159	Carnitine biosynthesis in mammals. *Biochemical Journal*, **2002**, 361, 417-29	3.8	286
158	Cardiolipin provides an essential activating platform for caspase-8 on mitochondria. *Journal of Cell Biology*, **2008**, 183, 681-96	7.3	229
157	X-linked cardioskeletal myopathy and neutropenia (Barth syndrome): an update. *American Journal of Medical Genetics Part A*, **2004**, 126A, 349-54		215
156	Cardiac and skeletal muscle defects in a mouse model of human Barth syndrome. *Journal of Biological Chemistry*, **2011**, 286, 899-908	5.4	170
155	Mutations in LPIN1 cause recurrent acute myoglobinuria in childhood. *American Journal of Human Genetics*, **2008**, 83, 489-94	11	165
154	Aberrant cardiolipin metabolism in the yeast taz1 mutant: a model for Barth syndrome. *Molecular Microbiology*, **2004**, 51, 149-58	4.1	164
153	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	36.3	147
152	Sodium taurocholate cotransporting polypeptide (SLC10A1) deficiency: conjugated hypercholanemia without a clear clinical phenotype. *Hepatology*, **2015**, 61, 260-7	11.2	130
151	The enigmatic role of tafazzin in cardiolipin metabolism. *Biochimica Et Biophysica Acta - Biomembranes*, **2009**, 1788, 2003-14	3.8	117
150	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, 1194-206	6.9	113
149	Cardiolipin deficiency affects respiratory chain function and organization in an induced pluripotent stem cell model of Barth syndrome. *Stem Cell Research*, **2013**, 11, 806-19	1.6	110
148	Monolysocardiolipins accumulate in Barth syndrome but do not lead to enhanced apoptosis. *Journal of Lipid Research*, **2005**, 46, 1182-95	6.3	108
147	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, 43089-94	5.4	101
146	LPIN1 gene mutations: a major cause of severe rhabdomyolysis in early childhood. *Human Mutation*, **2010**, 31, E1564-73	4.7	100

145	Cardiolipin and monolysocardiolipin analysis in fibroblasts, lymphocytes, and tissues using high-performance liquid chromatography-mass spectrometry as a diagnostic test for Barth syndrome. *Analytical Biochemistry*, **2009**, 387, 230-7	3.1	97
144	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, 7974-81	11.5	94
143	Bloodspot assay using HPLC-tandem mass spectrometry for detection of Barth syndrome. *Clinical Chemistry*, **2008**, 54, 371-8	5.5	90
142	The human TAZ gene complements mitochondrial dysfunction in the yeast taz1Delta mutant. Implications for Barth syndrome. *Journal of Biological Chemistry*, **2004**, 279, 44394-9	5.4	85
141	CLPB mutations cause 3-methylglutaconic aciduria, progressive brain atrophy, intellectual disability, congenital neutropenia, cataracts, movement disorder. *American Journal of Human Genetics*, **2015**, 96, 245-57	11	84
140	PPAR alpha-activation results in enhanced carnitine biosynthesis and OCTN2-mediated hepatic carnitine accumulation. *Biochimica Et Biophysica Acta - Bioenergetics*, **2007**, 1767, 1134-42	4.6	74
139	Identification and characterization of human cardiolipin synthase. *FEBS Letters*, **2006**, 580, 3059-64	3.8	74
138	Characterization of carnitine and fatty acid metabolism in the long-chain acyl-CoA dehydrogenase-deficient mouse. *Biochemical Journal*, **2005**, 387, 185-93	3.8	73
137	Bile acid analysis in human disorders of bile acid biosynthesis. *Molecular Aspects of Medicine*, **2017**, 56, 10-24	16.7	64
136	Molecular and biochemical characterization of rat gamma-trimethylaminobutyraldehyde dehydrogenase and evidence for the involvement of human aldehyde dehydrogenase 9 in carnitine biosynthesis. *Journal of Biological Chemistry*, **2000**, 275, 7390-4	5.4	58
135	Zellweger spectrum disorders: clinical manifestations in patients surviving into adulthood. *Journal of Inherited Metabolic Disease*, **2016**, 39, 93-106	5.4	56
134	Enzymology of the carnitine biosynthesis pathway. *IUBMB Life*, **2010**, 62, 357-62	4.7	56
133	Growth and fatty acid profiles of VLBW infants receiving a multicomponent lipid emulsion from birth. *Journal of Pediatric Gastroenterology and Nutrition*, **2014**, 58, 417-27	2.8	55
132	Distinct effects of tafazzin deletion in differentiated and undifferentiated mitochondria. *Mitochondrion*, **2009**, 9, 86-95	4.9	55
131	Clinical and biochemical characterization of four patients with mutations in ECHS1. *Orphanet Journal of Rare Diseases*, **2015**, 10, 79	4.2	51
130	Retrospective study of long-term outcomes of enzyme replacement therapy in Fabry disease: Analysis of prognostic factors. *PLoS ONE*, **2017**, 12, e0182379	3.7	51
129	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	3.7	46
128	Lipoprotein Lipase Maintains Microglial Innate Immunity in Obesity. *Cell Reports*, **2017**, 20, 3034-3042	10.6	46

127	Deletion of the cardiolipin-specific phospholipase Cld1 rescues growth and life span defects in the tafazzin mutant: implications for Barth syndrome. *Journal of Biological Chemistry*, **2014**, 289, 3114-25	5.4	44
126	Molecular and Biochemical Characterization of Rat epsilon -N-Trimethyllysine Hydroxylase, the First Enzyme of Carnitine Biosynthesis. *Journal of Biological Chemistry*, **2001**, 276, 33512-7	5.4	44
125	Carnitine biosynthesis: identification of the cDNA encoding human gamma-butyrobetaine hydroxylase. *Biochemical and Biophysical Research Communications*, **1998**, 250, 506-10	3.4	44
124	A sensitive mass spectrometry platform identifies metabolic changes of life history traits in C. elegans. *Scientific Reports*, **2017**, 7, 2408	4.9	41
123	Agalsidase alfa versus agalsidase beta for the treatment of Fabry disease: an international cohort study. *Journal of Medical Genetics*, **2018**, 55, 351-358	5.8	41
122	Mutations in PCYT2 disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia. *Brain*, **2019**, 142, 3382-3397	11.2	40
121	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	3.7	38
120	Lipidomic analysis of fibroblasts from Zellweger spectrum disorder patients identifies disease-specific phospholipid ratios. *Journal of Lipid Research*, **2016**, 57, 1447-54	6.3	38
119	Delineating the role of alterations in lipid metabolism to the pathogenesis of inherited skeletal and cardiac muscle disorders: Thematic Review Series: Genetics of Human Lipid Diseases. *Journal of Lipid Research*, **2012**, 53, 4-27	6.3	38
118	Defining functional classes of Barth syndrome mutation in humans. *Human Molecular Genetics*, **2016**, 25, 1754-70	5.6	37
117	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	6.9	37
116	Tissue carnitine homeostasis in very-long-chain acyl-CoA dehydrogenase-deficient mice. *Pediatric Research*, **2005**, 57, 760-4	3.2	36
115	Peroxisomes can oxidize medium- and long-chain fatty acids through a pathway involving ABCD3 and HSD17B4. *FASEB Journal*, **2019**, 33, 4355-4364	0.9	36
114	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	7.5	35
113	The important role of biochemical and functional studies in the diagnostics of peroxisomal disorders. *Journal of Inherited Metabolic Disease*, **2016**, 39, 531-43	5.4	34
112	Cholic acid therapy in Zellweger spectrum disorders. *Journal of Inherited Metabolic Disease*, **2016**, 39, 859-868	5.4	33
111	The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome. *Orphanet Journal of Rare Diseases*, **2017**, 12, 49	4.2	32
110	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	4.9	32

109	Functional characterisation of peroxisomal β-oxidation disorders in fibroblasts using lipidomics. *Journal of Inherited Metabolic Disease*, **2018**, 41, 479-487	5.4	30
108	Measurement of carnitine biosynthesis enzyme activities by tandem mass spectrometry: differences between the mouse and the rat. *Analytical Biochemistry*, **2006**, 354, 132-9	3.1	30
107	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	6.9	29
106	Principles and practice of lipidomics. *Journal of Inherited Metabolic Disease*, **2015**, 38, 41-52	5.4	29
105	Organic solute transporter-α/β (SLC51B) deficiency in two brothers with congenital diarrhea and features of cholestasis. *Hepatology*, **2018**, 68, 590-598	11.2	29
104	Fatty acid metabolism and its longitudinal relationship with the hypothalamic-pituitary-adrenal axis in major depression: Associations with prospective antidepressant response. *Psychoneuroendocrinology*, **2015**, 59, 1-13	5	28
103	Identification and characterization of a complete carnitine biosynthesis pathway in Candida albicans. *FASEB Journal*, **2009**, 23, 2349-59	0.9	28
102	C26:0-Carnitine Is a New Biomarker for X-Linked Adrenoleukodystrophy in Mice and Man. *PLoS ONE* , **2016**, 11, e0154597	3.7	28
101	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	3.7	26
100	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	3.8	26
99	New clinical and molecular insights on Barth syndrome. *Orphanet Journal of Rare Diseases*, **2013**, 8, 27	4.2	26
98	New targets for monitoring and therapy in Barth syndrome. *Genetics in Medicine*, **2016**, 18, 1001-10	8.1	25
97	Evaluation of C26:0-lysophosphatidylcholine and C26:0-carnitine as diagnostic markers for Zellweger spectrum disorders. *Journal of Inherited Metabolic Disease*, **2017**, 40, 875-881	5.4	25
96	Hepatotoxicity due to chenodeoxycholic acid supplementation in an infant with cerebrotendinous xanthomatosis: implications for treatment. *European Journal of Pediatrics*, **2016**, 175, 143-6	4.1	24
95	Vulnerability for new episodes in recurrent major depressive disorder: protocol for the longitudinal DELTA-neuroimaging cohort study. *BMJ Open*, **2016**, 6, e009510	3	23
94	The dynamics of cardiolipin synthesis post-mitochondrial fusion. *Biochimica Et Biophysica Acta - Biomembranes*, **2010**, 1798, 1577-85	3.8	22
93	Adrenoleukodystrophy Newborn Screening in the Netherlands (SCAN Study): The X-Factor. *Frontiers in Cell and Developmental Biology*, **2020**, 8, 499	5.7	21
92	SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family. *Journal of Medical Genetics*, **2018**, 55, 39-47	5.8	21

91	The cellular and molecular mechanisms for neutropenia in Barth syndrome. *European Journal of Haematology*, **2012**, 88, 195-209	3.8	20
90	Carnitine biosynthesis in Neurospora crassa: identification of a cDNA coding for epsilon-N-trimethyllysine hydroxylase and its functional expression in Saccharomyces cerevisiae. *FEMS Microbiology Letters*, **2002**, 210, 19-23	2.9	20
89	NTCP deficiency and persistently raised bile salts: an adult case. *Journal of Inherited Metabolic Disease*, **2017**, 40, 313-315	5.4	19
88	Levels of Red Blood Cell Fatty Acids in Patients With Psychosis, Their Unaffected Siblings, and Healthy Controls. *Schizophrenia Bulletin*, **2016**, 42, 358-68	1.3	19
87	A newborn screening method for cerebrotendinous xanthomatosis using bile alcohol glucuronides and metabolite ratios. *Journal of Lipid Research*, **2017**, 58, 1002-1007	6.3	18
86	Skeletal muscle in healthy humans exhibits a day-night rhythm in lipid metabolism. *Molecular Metabolism*, **2020**, 37, 100989	8.8	18
85	Cardiolipin molecular species with shorter acyl chains accumulate in Saccharomyces cerevisiae mutants lacking the acyl coenzyme A-binding protein Acb1p: new insights into acyl chain remodeling of cardiolipin. *Journal of Biological Chemistry*, **2009**, 284, 27609-19	5.4	18
84	An improved enzyme assay for carnitine palmitoyl transferase I in fibroblasts using tandem mass spectrometry. *Molecular Genetics and Metabolism*, **2007**, 90, 24-9	3.7	18
83	Mitochondrial Encephalopathy and Transient 3-Methylglutaconic Aciduria in ECHS1 Deficiency: Long-Term Follow-Up. *JIMD Reports*, **2018**, 39, 83-87	1.9	17
82	A mutation creating an upstream translation initiation codon in SLC22A5 5′UTR is a frequent cause of primary carnitine deficiency. *Human Mutation*, **2019**, 40, 1899-1904	4.7	16
81	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, 11-5	1.8	16
80	Plasma lipidomics as a diagnostic tool for peroxisomal disorders. *Journal of Inherited Metabolic Disease*, **2018**, 41, 489-498	5.4	14
79	Ethnic differences in metabolite signatures and type 2 diabetes: a nested case-control analysis among people of South Asian, African and European origin. *Nutrition and Diabetes*, **2017**, 7, 300	4.7	14
78	Urinary metabolic phenotyping of mucopolysaccharidosis type I combining untargeted and targeted strategies with data modeling. *Clinica Chimica Acta*, **2017**, 475, 7-14	6.2	13
77	Functional analysis of TMLH variants and definition of domains required for catalytic activity and mitochondrial targeting. *Journal of Cellular Physiology*, **2005**, 204, 839-47	7	13
76	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, 1375-82	6.9	13
75	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	5.4	13
74	Submitochondrial localization of 6-N-trimethyllysine dioxygenase - implications for carnitine biosynthesis. *FEBS Journal*, **2007**, 274, 5845-51	5.7	12

73	The cholic acid extension study in Zellweger spectrum disorders: Results and implications for therapy. *Journal of Inherited Metabolic Disease*, **2019**, 42, 303-312	5.4	11
72	Supplementation with a powdered blend of PUFAs normalizes DHA and AA levels in patients with PKU. *Molecular Genetics and Metabolism*, **2013**, 109, 121-4	3.7	11
71	Lipidomics unveils lipid dyshomeostasis and low circulating plasmalogens as biomarkers in a monogenic mitochondrial disorder. *JCI Insight*, **2019**, 4,	9.9	11
70	Mitochondrial Fatty Acid Oxidation Disorders: Laboratory Diagnosis, Pathogenesis, and the Complicated Route to Treatment. *Journal of Lipid and Atherosclerosis*, **2020**, 9, 313-333	3	11
69	Mitochondrial disruption in peroxisome deficient cells is hepatocyte selective but is not mediated by common hepatic peroxisomal metabolites. *Mitochondrion*, **2018**, 39, 51-59	4.9	11
68	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	5.7	11
67	Analysis of carnitine biosynthesis metabolites in urine by HPLC-electrospray tandem mass spectrometry. *Clinical Chemistry*, **2002**, 48, 826-34	5.5	11
66	Polyunsaturated fatty acid biostatus, phospholipase A activity and brain white matter microstructure across adolescence. *Neuroscience*, **2017**, 343, 423-433	3.9	10
65	Bile Acid Alters Male Mouse Fertility in Metabolic Syndrome Context. *PLoS ONE*, **2015**, 10, e0139946	3.7	10
64	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,	4.5	9
63	Disturbed brain ether lipid metabolism and histology in Sjögren-Larsson syndrome. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1265-1278	5.4	9
62	The effect of mirabegron on energy expenditure and brown adipose tissue in healthy lean South Asian and Europid men. *Diabetes, Obesity and Metabolism*, **2020**, 22, 2032-2044	6.7	9
61	Carnitine Biosynthesis. *Advances in Experimental Medicine and Biology*, **2002**, 117-124	3.6	9
60	Fatty Acid Oxidation in Peroxisomes: Enzymology, Metabolic Crosstalk with Other Organelles and Peroxisomal Disorders. *Advances in Experimental Medicine and Biology*, **2020**, 1299, 55-70	3.6	9
59	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	8.1	8
58	Plasma oxalate: comparison of methodologies. *Urolithiasis*, **2020**, 48, 473-480	3.2	8
57	Multi-Omic Approach to Identify Phenotypic Modifiers Underlying Cerebral Demyelination in X-Linked Adrenoleukodystrophy. *Frontiers in Cell and Developmental Biology*, **2020**, 8, 520	5.7	8
56	Reliable Diagnosis of Carnitine Palmitoyltransferase Type IA Deficiency by Analysis of Plasma Acylcarnitine Profiles. *JIMD Reports*, **2017**, 32, 33-39	1.9	8

55	Mouse Tafazzin Is Required for Male Germ Cell Meiosis and Spermatogenesis. *PLoS ONE*, **2015**, 10, e0131066	3.7	8
54	Lipidomics in Nonalcoholic Fatty Liver Disease: Exploring Serum Lipids as Biomarkers for Pediatric Nonalcoholic Fatty Liver Disease. *Journal of Pediatric Gastroenterology and Nutrition*, **2020**, 71, 433-439	2.8	8
53	Identification of Metabolic Biomarkers in Relation to Methotrexate Response in Early Rheumatoid Arthritis. *Journal of Personalized Medicine*, **2020**, 10,	3.6	8
52	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	11.5	8
51	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	2	7
50	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	3.2	7
49	Biochemical and imaging parameters in acid sphingomyelinase deficiency: Potential utility as biomarkers. *Molecular Genetics and Metabolism*, **2020**, 130, 16-26	3.7	7
48	Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation?. *JIMD Reports*, **2016**, 29, 89-93	1.9	7
47	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	8.1	7
46	Inherited disorders of complex lipid metabolism: A clinical review. *Journal of Inherited Metabolic Disease*, **2021**, 44, 809-825	5.4	6
45	Oral therapy for riboflavin transporter deficiency - What is the regimen of choice?. *Parkinsonism and Related Disorders*, **2019**, 61, 245-247	3.6	6
44	Slc22a5 haploinsufficiency does not aggravate the phenotype of the long-chain acyl-CoA dehydrogenase KO mouse. *Journal of Inherited Metabolic Disease*, **2020**, 43, 486-495	5.4	5
43	Cerebellar and hepatic alterations in ACBD5-deficient mice are associated with unexpected, distinct alterations in cellular lipid homeostasis. *Communications Biology*, **2020**, 3, 713	6.7	5
42	Metabolomics and lipidomics in Caenorhabditis elegans using a single-sample preparation. *DMM Disease Models and Mechanisms*, **2021**, 14,	4.1	5
41	Intra-individual plasticity of the TAZ gene leading to different heritable mutations in siblings with Barth syndrome. *European Journal of Human Genetics*, **2015**, 23, 1708-12	5.3	4
40	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	6	4
39	Indoleamine 2,3-dioxygenase (IDO)-1 and IDO-2 activity and severe course of COVID-19. *Journal of Pathology*, **2021**,	9.4	4
38	Aging selectively dampens oscillation of lipid abundance in white and brown adipose tissue. *Scientific Reports*, **2021**, 11, 5932	4.9	4

37	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	5.7	4
36	Biochemical Screening of Intellectually Disabled Patients: A Stepping Stone to Initiate a Newborn Screening Program in Pakistan. *Frontiers in Neurology*, **2019**, 10, 762	4.1	3
35	Identification and diagnostic value of phytanoyl- and pristanoyl-carnitine in plasma from patients with peroxisomal disorders. *Molecular Genetics and Metabolism*, **2017**, 121, 279-282	3.7	3
34	Cell Type-Selective Loss of Peroxisomal β-Oxidation Impairs Bipolar Cell but Not Photoreceptor Survival in the Retina.. *Cells*, **2022**, 11,	7.9	3
33	Mild Zellweger syndrome due to functionally confirmed novel PEX1 variants. *Journal of Applied Genetics*, **2020**, 61, 87-91	2.5	3
32	ECHS1 disease in two unrelated families of Samoan descent: Common variant - rare disorder. *American Journal of Medical Genetics, Part A*, **2021**, 185, 157-167	2.5	3
31	Enantiomer-specific pharmacokinetics of D,L-3-hydroxybutyrate: Implications for the treatment of multiple acyl-CoA dehydrogenase deficiency. *Journal of Inherited Metabolic Disease*, **2021**, 44, 926-938	5.4	3
30	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	5.6	3
29	PTPMT1 Is Required for Embryonic Cardiac Cardiolipin Biosynthesis to Regulate Mitochondrial Morphogenesis and Heart Development. *Circulation*, **2021**, 144, 403-406	16.7	3
28	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	5.4	3
27	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	3.7	2
26	Biochemical Studies in Fibroblasts to Interpret Variants of Unknown Significance in the Gene.. *Genes*, **2021**, 12,	4.2	2
25	CHAPTER 13:Dioxygenases of Carnitine Biosynthesis: 6-N-Trimethyllysine and γ-Butyrobetaine Hydroxylases. *2-Oxoglutarate-Dependent Oxygenases*, **2015**, 324-337	1.8	2
24	Delayed appearance of 3-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	2.5	2
23	Cardiolipin Remodeling Defects Impair Mitochondrial Architecture and Function in a Murine Model of Barth Syndrome Cardiomyopathy. *Circulation: Heart Failure*, **2021**, 14, e008289	7.6	2
22	Exploring the metabolic fate of medium-chain triglycerides in healthy individuals using a stable isotope tracer. *Clinical Nutrition*, **2021**, 40, 1396-1404	5.9	2
21	The cholic acid extension study in Zellweger spectrum disorders: results and implications for therapy. *Journal of Inherited Metabolic Disease*, **2018**,	5.4	2
20	Circadian misalignment disturbs the skeletal muscle lipidome in healthy young men. *FASEB Journal*, **2021**, 35, e21611	0.9	1

19	Meijer and Vloedman's histochemical demonstration of mitochondrial coupling obeys Lambert-Beer's law in the myocardium. *Histochemistry and Cell Biology*, **2019**, 151, 85-90	2.4	1
18	Monitoring phenylalanine concentrations in the follow-up of phenylketonuria patients: An inventory of pre-analytical and analytical variation. *JIMD Reports*, **2021**, 58, 70-79	1.9	1
17	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	11	1
16	Reduced ech-6 expression attenuates fat-induced lifespan shortening in C. elegans.. *Scientific Reports*, **2022**, 12, 3350	4.9	1
15	Biallelic variants in are associated with low muscle cardiolipin levels, leading to neonatal mitochondrial disease.. *Human Genetics and Genomics Advances*, **2022**, 3, 100097	0.8	1
14	Mice with a deficiency in Peroxisomal Membrane Protein 4 (PXMP4) display mild changes in hepatic lipid metabolism.. *Scientific Reports*, **2022**, 12, 2512	4.9	1
13	Inborn Errors of Non-Mitochondrial Fatty Acid Metabolism Including Peroxisomal Disorders **2022**, 785-809		1
12	Time-restricted feeding during the inactive phase abolishes the daily rhythm in mitochondrial respiration in rat skeletal muscle.. *FASEB Journal*, **2022**, 36, e22133	0.9	0
11	Neonatal carnitine concentrations in relation to gestational age and weight. *JIMD Reports*, **2020**, 56, 95-104	1.9	0
10	Reply: Expanding the clinical and genetic spectrum of PCYT2-related disorders. *Brain*, **2020**, 143, e77	11.2	0
9	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, 2326409818810[illegible]28	1.4	0
8	An improved functional assay in blood spot to diagnose Barth syndrome using the monolysocardiolipin/cardiolipin ratio. *Journal of Inherited Metabolic Disease*, **2021**, 45, 29	5.4	0
7	Polar metabolomics in human muscle biopsies using a liquid-liquid extraction and full-scan LC-MS.. *STAR Protocols*, **2022**, 3, 101302	1.4	0
6	A lipidomics approach to identify disorders in phospholipid metabolism; MEGDEL syndrome unraveled. *Tijdschrift Voor Kindergeneeskunde*, **2013**, 81, 112-112		
5	Topological Difference but Dysfunctional Conservation of Cardiolipin Remodeling in Yeast and Mammals. *FASEB Journal*, **2015**, 29, 885.12	0.9	
4	The Cellular and Molecular Mechanisms of Neutropenia in Barth Syndrome. *Blood*, **2011**, 118, 1105-1105	2.2	
3	Disorders of Complex Lipids **2022**, 981-1025		
2	Disorders of Bile Acid Synthesis **2022**, 1095-1112		

1	Adaptations of the 3T3-L1 adipocyte lipidome to defective ether lipid catabolism upon alkylglycerol monooxygenase knockdown.. *Journal of Lipid Research*, **2022**, 100222	6.3