Jerry Vockley

List of Publications by Citations

281 papers	9,524 citations	56 h-index	85 g-index
311 ext. papers	11,362 ext. citations	4.6 avg, IF	5.92 L-index

#	Paper	IF	Citations
281	Increased levels of plasma acylcarnitines in obesity and type 2 diabetes and identification of a marker of glucolipotoxicity. *Obesity*, **2010**, 18, 1695-700	8	392
280	Effects of diet and physical activity interventions on weight loss and cardiometabolic risk factors in severely obese adults: a randomized trial. *JAMA - Journal of the American Medical Association*, **2010**, 304, 1795-802	27.4	353
279	Phenylalanine hydroxylase deficiency: diagnosis and management guideline. *Genetics in Medicine*, **2014**, 16, 188-200	8.1	326
278	Targeted disruption of mouse long-chain acyl-CoA dehydrogenase gene reveals crucial roles for fatty acid oxidation. *Proceedings of the National Academy of Sciences of the United States of America*, **1998**, 95, 15592-7	11.5	185
277	Metabolomic profiling of fatty acid and amino acid metabolism in youth with obesity and type 2 diabetes: evidence for enhanced mitochondrial oxidation. *Diabetes Care*, **2012**, 35, 605-11	14.6	182
276	Synergistic heterozygosity: disease resulting from multiple partial defects in one or more metabolic pathways. *Molecular Genetics and Metabolism*, **2000**, 71, 10-8	3.7	176
275	Isovaleric acidemia: new aspects of genetic and phenotypic heterogeneity. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2006**, 142C, 95-103	3.1	144
274	Efficacy of sapropterin dihydrochloride in increasing phenylalanine tolerance in children with phenylketonuria: a phase III, randomized, double-blind, placebo-controlled study. *Journal of Pediatrics*, **2009**, 154, 700-7	3.6	142
273	Molecular heterogeneity in very-long-chain acyl-CoA dehydrogenase deficiency causing pediatric cardiomyopathy and sudden death. *Circulation*, **1999**, 99, 1337-43	16.7	135
272	Sirtuin 3 (SIRT3) protein regulates long-chain acyl-CoA dehydrogenase by deacetylating conserved lysines near the active site. *Journal of Biological Chemistry*, **2013**, 288, 33837-33847	5.4	123
271	Barriers to the successful treatment of liver disease by hepatocyte transplantation. *Journal of Hepatology*, **2010**, 53, 769-74	13.4	122
270	A common mutation is associated with a mild, potentially asymptomatic phenotype in patients with isovaleric acidemia diagnosed by newborn screening. *American Journal of Human Genetics*, **2004**, 75, 1136-42	11	119
269	Recommendations for the nutrition management of phenylalanine hydroxylase deficiency. *Genetics in Medicine*, **2014**, 16, 121-31	8.1	117
268	Liver transplantation for classical maple syrup urine disease: long-term follow-up in 37 patients and comparative United Network for Organ Sharing experience. *Journal of Pediatrics*, **2012**, 160, 116-21.e1	3.6	116
267	Molecular Cloning and Nucleotide Sequence of cDNAs Encoding the Precursors of Rat Long Chain Acyl-Coenzyme A, Short Chain Acyl-Coenzyme A, and Isovaleryl-Coenzyme A Dehydrogenases. *Journal of Biological Chemistry*, **1989**, 264, 16321-16331	5.4	114
266	Molecular cloning and nucleotide sequence of cDNAs encoding the precursors of rat long chain acyl-coenzyme A, short chain acyl-coenzyme A, and isovaleryl-coenzyme A dehydrogenases. Sequence homology of four enzymes of the acyl-CoA dehydrogenase family. *Journal of Biological Chemistry*, **1989**, 264, 16321-31	5.4	112
265	Single-dose, subcutaneous recombinant phenylalanine ammonia lyase conjugated with polyethylene glycol in adult patients with phenylketonuria: an open-label, multicentre, phase 1 dose-escalation trial. *Lancet, The*, **2014**, 384, 37-44	40	109

264	Evidence for physical association of mitochondrial fatty acid oxidation and oxidative phosphorylation complexes. *Journal of Biological Chemistry*, **2010**, 285, 29834-41	5.4	109
263	Role of common gene variations in the molecular pathogenesis of short-chain acyl-CoA dehydrogenase deficiency. *Pediatric Research*, **2001**, 49, 18-23	3.2	105
262	Gestational, pathologic and biochemical differences between very long-chain acyl-CoA dehydrogenase deficiency and long-chain acyl-CoA dehydrogenase deficiency in the mouse. *Human Molecular Genetics*, **2001**, 10, 2069-77	5.6	99
261	A new genetic disorder in mitochondrial fatty acid beta-oxidation: ACAD9 deficiency. *American Journal of Human Genetics*, **2007**, 81, 87-103	11	96
260	A proposed nosology of inborn errors of metabolism. *Genetics in Medicine*, **2019**, 21, 102-106	8.1	92
259	Structure of human isovaleryl-CoA dehydrogenase at 2.6 A resolution: structural basis for substrate specificity,. *Biochemistry*, **1997**, 36, 8455-64	3.2	91
258	The ACADS gene variation spectrum in 114 patients with short-chain acyl-CoA dehydrogenase (SCAD) deficiency is dominated by missense variations leading to protein misfolding at the cellular level. *Human Genetics*, **2008**, 124, 43-56	6.3	84
257	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. *American Journal of Human Genetics*, **2015**, 97, 163-9	11	83
256	Liver transplantation for pediatric metabolic disease. *Molecular Genetics and Metabolism*, **2014**, 111, 418-27	3.7	82
255	Medium-chain acyl-CoA dehydrogenase deficiency in gene-targeted mice. *PLoS Genetics*, **2005**, 1, e23	6	82
254	Heritable connective tissue disorders in cervical artery dissections: a prospective study. *Neurology*, **1998**, 50, 1166-9	6.5	82
253	Randomized dose-escalation trial of elamipretide in adults with primary mitochondrial myopathy. *Neurology*, **2018**, 90, e1212-e1221	6.5	81
252	Mitochondrial structure, function and dynamics are temporally controlled by c-Myc. *PLoS ONE*, **2012** , 7, e37699	3.7	79
251	2-Methylbutyryl-coenzyme A dehydrogenase deficiency: a new inborn error of L-isoleucine metabolism. *Pediatric Research*, **2000**, 47, 830-3	3.2	79
250	Human acyl-CoA dehydrogenase-9 plays a novel role in the mitochondrial beta-oxidation of unsaturated fatty acids. *Journal of Biological Chemistry*, **2005**, 280, 32309-16	5.4	76
249	Pegvaliase for the treatment of phenylketonuria: Results of a long-term phase 3 clinical trial program (PRISM). *Molecular Genetics and Metabolism*, **2018**, 124, 27-38	3.7	75
248	Acute fatty liver of pregnancy associated with short-chain acyl-coenzyme A dehydrogenase deficiency. *Journal of Pediatrics*, **2001**, 138, 585-8	3.6	75
247	Severe hypoglycemia as a presenting symptom of carbohydrate-deficient glycoprotein syndrome. *Journal of Pediatrics*, **1999**, 135, 775-81	3.6	74

246	Mutations in the human SC4MOL gene encoding a methyl sterol oxidase cause psoriasiform dermatitis, microcephaly, and developmental delay. *Journal of Clinical Investigation*, **2011**, 121, 976-84	15.9	72
245	Rare disorders of metabolism with elevated butyryl- and isobutyryl-carnitine detected by tandem mass spectrometry newborn screening. *Pediatric Research*, **2003**, 54, 219-23	3.2	71
244	Defect in Dimethylglycine Dehydrogenase, a New Inborn Error of Metabolism: NMR Spectroscopy Study. *Clinical Chemistry*, **1999**, 45, 459-464	5.5	71
243	Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. *Journal of Inherited Metabolic Disease*, **2016**, 39, 3-16	5.4	68
242	Structural basis for substrate fatty acyl chain specificity: crystal structure of human very-long-chain acyl-CoA dehydrogenase. *Journal of Biological Chemistry*, **2008**, 283, 9435-43	5.4	68
241	Acyl-CoA dehydrogenases: Dynamic history of protein family evolution. *Journal of Molecular Evolution*, **2009**, 69, 176-93	3.1	67
240	Misfolding, degradation, and aggregation of variant proteins. The molecular pathogenesis of short chain acyl-CoA dehydrogenase (SCAD) deficiency. *Journal of Biological Chemistry*, **2003**, 278, 47449-58	5.4	67
239	c-Myc programs fatty acid metabolism and dictates acetyl-CoA abundance and fate. *Journal of Biological Chemistry*, **2014**, 289, 25382-92	5.4	66
238	Long-term major clinical outcomes in patients with long chain fatty acid oxidation disorders before and after transition to triheptanoin treatment--A retrospective chart review. *Molecular Genetics and Metabolism*, **2015**, 116, 53-60	3.7	65
237	Defects of mitochondrial beta-oxidation: a growing group of disorders. *Neuromuscular Disorders*, **2002**, 12, 235-46	2.9	65
236	Identification and characterization of new long chain acyl-CoA dehydrogenases. *Molecular Genetics and Metabolism*, **2011**, 102, 418-29	3.7	63
235	Short-chain acyl-coenzyme A dehydrogenase deficiency. *Molecular Genetics and Metabolism*, **2008**, 95, 195-200	3.7	63
234	Clinical and neurocognitive outcome in symptomatic isovaleric acidemia. *Orphanet Journal of Rare Diseases*, **2012**, 7, 9	4.2	62
233	Crystal structure of rat short chain acyl-CoA dehydrogenase complexed with acetoacetyl-CoA: comparison with other acyl-CoA dehydrogenases. *Journal of Biological Chemistry*, **2002**, 277, 12200-7	5.4	61
232	Long-chain acyl-CoA dehydrogenase is a key enzyme in the mitochondrial beta-oxidation of unsaturated fatty acids. *Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids*, **2000**, 1485, 121-8	5	61
231	Short-chain acyl-CoA dehydrogenase deficiency: a cause of ophthalmoplegia and multicore myopathy. *Neurology*, **1999**, 52, 366-72	6.5	61
230	Cloning of dimethylglycine dehydrogenase and a new human inborn error of metabolism, dimethylglycine dehydrogenase deficiency. *American Journal of Human Genetics*, **2001**, 68, 839-47	11	59
229	Metabolic Reprogramming in Astrocytes Distinguishes Region-Specific Neuronal Susceptibility in Huntington Mice. *Cell Metabolism*, **2019**, 29, 1258-1273.e11	24.6	58

228	Triheptanoin versus trioctanoin for long-chain fatty acid oxidation disorders: a double blinded, randomized controlled trial. *Journal of Inherited Metabolic Disease*, **2017**, 40, 831-843	5.4	57
227	Metabolomic profiling of amino acids and β-cell function relative to insulin sensitivity in youth. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E2119-24	5.6	56
226	Advances and challenges in the treatment of branched-chain amino/keto acid metabolic defects. *Journal of Inherited Metabolic Disease*, **2012**, 35, 29-40	5.4	54
225	Isolation and expression of a cDNA encoding the precursor for a novel member (ACADSB) of the acyl-CoA dehydrogenase gene family. *Genomics*, **1994**, 24, 280-7	4.3	54
224	Characterization of molecular defects in isovaleryl-CoA dehydrogenase in patients with isovaleric acidemia. *Biochemistry*, **1998**, 37, 10325-35	3.2	53
223	Short-chain acyl-CoA dehydrogenase gene mutation (c.319C>T) presents with clinical heterogeneity and is candidate founder mutation in individuals of Ashkenazi Jewish origin. *Molecular Genetics and Metabolism*, **2008**, 93, 179-89	3.7	53
222	Prospective diagnosis of 2-methylbutyryl-CoA dehydrogenase deficiency in the Hmong population by newborn screening using tandem mass spectrometry. *Pediatrics*, **2003**, 112, 74-8	7.4	53
221	Identification of isobutyryl-CoA dehydrogenase and its deficiency in humans. *Molecular Genetics and Metabolism*, **2002**, 77, 68-79	3.7	52
220	Outcomes and genotype-phenotype correlations in 52 individuals with VLCAD deficiency diagnosed by NBS and enrolled in the IBEM-IS database. *Molecular Genetics and Metabolism*, **2016**, 118, 272-81	3.7	51
219	Exon skipping in IVD RNA processing in isovaleric acidemia caused by point mutations in the coding region of the IVD gene. *American Journal of Human Genetics*, **2000**, 66, 356-67	11	51
218	Structures of isobutyryl-CoA dehydrogenase and enzyme-product complex: comparison with isovaleryl- and short-chain acyl-CoA dehydrogenases. *Journal of Biological Chemistry*, **2004**, 279, 16526-34	5.4	50
217	Synergistic heterozygosity in mice with inherited enzyme deficiencies of mitochondrial fatty acid beta-oxidation. *Molecular Genetics and Metabolism*, **2005**, 85, 7-11	3.7	48
216	Triheptanoin treatment in patients with pediatric cardiomyopathy associated with long chain-fatty acid oxidation disorders. *Molecular Genetics and Metabolism*, **2016**, 119, 223-231	3.7	47
215	UX007 for the treatment of long chain-fatty acid oxidation disorders: Safety and efficacy in children and adults following 24weeks of treatment. *Molecular Genetics and Metabolism*, **2017**, 120, 370-377	3.7	44
214	Complex I assembly function and fatty acid oxidation enzyme activity of ACAD9 both contribute to disease severity in ACAD9 deficiency. *Human Molecular Genetics*, **2015**, 24, 3238-47	5.6	44
213	A novel mitochondrial tRNA(Leu(UUR)) mutation in a patient with features of MERRF and Kearns-Sayre syndrome. *Neuromuscular Disorders*, **2003**, 13, 334-40	2.9	44
212	Identification of the active site catalytic residue in human isovaleryl-CoA dehydrogenase. *Biochemistry*, **1995**, 34, 10146-52	3.2	44
211	The Genetic Landscape and Epidemiology of Phenylketonuria. *American Journal of Human Genetics*, **2020**, 107, 234-250	11	44

210	Cognitive and adaptive functioning after liver transplantation for maple syrup urine disease: a case series. *Pediatric Transplantation*, **2011**, 15, 58-64	1.8	42
209	High-level expression of an altered cDNA encoding human isovaleryl-CoA dehydrogenase in Escherichia coli. *Gene*, **1995**, 160, 263-7	3.8	42
208	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. *Orphanet Journal of Rare Diseases*, **2018**, 13, 120	4.2	41
207	Women have higher protein content of beta-oxidation enzymes in skeletal muscle than men. *PLoS ONE*, **2010**, 5, e12025	3.7	41
206	Development of a newborn screening follow-up algorithm for the diagnosis of isobutyryl-CoA dehydrogenase deficiency. *Genetics in Medicine*, **2007**, 9, 108-16	8.1	41
205	Liver Transplantation for Propionic Acidemia and Methylmalonic Acidemia: Perioperative Management and Clinical Outcomes. *Liver Transplantation*, **2018**, 24, 1260-1270	4.5	39
204	Impaired tetramer assembly of variant medium-chain acyl-coenzyme A dehydrogenase with a glutamate or aspartate substitution for lysine 304 causing instability of the protein. *Journal of Biological Chemistry*, **1992**, 267, 26004-10	5.4	39
203	Neurometabolic Disorders: Potentially Treatable Abnormalities in Patients With Treatment-Refractory Depression and Suicidal Behavior. *American Journal of Psychiatry*, **2017**, 174, 42-50	11.9	38
202	Molecular and cellular pathology of very-long-chain acyl-CoA dehydrogenase deficiency. *Molecular Genetics and Metabolism*, **2013**, 109, 21-7	3.7	37
201	Mitochondrial fatty acid oxidation and the electron transport chain comprise a multifunctional mitochondrial protein complex. *Journal of Biological Chemistry*, **2019**, 294, 12380-12391	5.4	36
200	Evaluation of mitochondrial bioenergetics, dynamics, endoplasmic reticulum-mitochondria crosstalk, and reactive oxygen species in fibroblasts from patients with complex I deficiency. *Scientific Reports*, **2018**, 8, 1165	4.9	36
199	Follistatin-like protein 1 enhances NLRP3 inflammasome-mediated IL-1β secretion from monocytes and macrophages. *European Journal of Immunology*, **2014**, 44, 1467-79	6.1	36
198	Expression and characterization of mutations in human very long-chain acyl-CoA dehydrogenase using a prokaryotic system. *Molecular Genetics and Metabolism*, **2007**, 91, 138-47	3.7	36
197	Diagnosis and management of defects of mitochondrial beta-oxidation. *Current Opinion in Clinical Nutrition and Metabolic Care*, **2002**, 5, 601-9	3.8	36
196	The changing face of disorders of fatty acid oxidation. *Mayo Clinic Proceedings*, **1994**, 69, 249-57	6.4	36
195	Impaired tetramer assembly of variant medium-chain acyl-coenzyme A dehydrogenase with a glutamate or aspartate substitution for lysine 304 causing instability of the protein.. *Journal of Biological Chemistry*, **1992**, 267, 26004-26010	5.4	35
194	Long-chain acyl-CoA dehydrogenase deficiency as a cause of pulmonary surfactant dysfunction. *Journal of Biological Chemistry*, **2014**, 289, 10668-10679	5.4	34
193	Metabolism as a complex genetic trait, a systems biology approach: implications for inborn errors of metabolism and clinical diseases. *Journal of Inherited Metabolic Disease*, **2008**, 31, 619-29	5.4	33

192	Nutritional interventions in primary mitochondrial disorders: Developing an evidence base. *Molecular Genetics and Metabolism*, **2016**, 119, 187-206	3.7	33
191	Pegvaliase for the treatment of phenylketonuria: A pivotal, double-blind randomized discontinuation Phase 3 clinical trial. *Molecular Genetics and Metabolism*, **2018**, 124, 20-26	3.7	32
190	An international classification of inherited metabolic disorders (ICIMD). *Journal of Inherited Metabolic Disease*, **2021**, 44, 164-177	5.4	32
189	Urinary phenylacetylglutamine as dosing biomarker for patients with urea cycle disorders. *Molecular Genetics and Metabolism*, **2012**, 107, 308-14	3.7	31
188	Efficacy and Safety of Asfotase Alfa in Infants and Young Children With Hypophosphatasia: A Phase 2 Open-Label Study. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2735-2747	5.6	30
187	Novel mutation in MYH7 gene associated with distal myopathy and cardiomyopathy. *Neuromuscular Disorders*, **2011**, 21, 219-22	2.9	30
186	Characterization of new ACADSB gene sequence mutations and clinical implications in patients with 2-methylbutyrylglycinuria identified by newborn screening. *Molecular Genetics and Metabolism* , **2010**, 100, 333-8	3.7	30
185	Enhanced defense against Pneumocystis carinii mediated by a novel dectin-1 receptor Fc fusion protein. *Journal of Immunology*, **2007**, 178, 3702-12	5.3	29
184	Cloning of a cDNA for short/branched chain acyl-Coenzyme A dehydrogenase from rat and characterization of its tissue expression and substrate specificity. *Archives of Biochemistry and Biophysics*, **1996**, 331, 127-33	4.1	29
183	Evidence- and consensus-based recommendations for the use of pegvaliase in adults with phenylketonuria. *Genetics in Medicine*, **2019**, 21, 1851-1867	8.1	29
182	Inborn Errors of Metabolism with Myopathy: Defects of Fatty Acid Oxidation and the Carnitine Shuttle System. *Pediatric Clinics of North America*, **2018**, 65, 317-335	3.6	28
181	Sapropterin dihydrochloride use in pregnant women with phenylketonuria: an interim report of the PKU MOMS sub-registry. *Molecular Genetics and Metabolism*, **2014**, 112, 9-16	3.7	28
180	Neuropsychological outcomes in fatty acid oxidation disorders: 85 cases detected by newborn screening. *Developmental Disabilities Research Reviews*, **2013**, 17, 260-8		28
179	Diagnosis of possible mitochondrial disease: an existential crisis. *Journal of Medical Genetics*, **2019**, 56, 123-130	5.8	27
178	Infantile hypophosphatasia secondary to a novel compound heterozygous mutation presenting with pyridoxine-responsive seizures. *JIMD Reports*, **2013**, 11, 17-24	1.9	27
177	Mammalian branched-chain acyl-CoA dehydrogenases: molecular cloning and characterization of recombinant enzymes. *Methods in Enzymology*, **2000**, 324, 241-58	1.7	27
176	Results from a 78-week, single-arm, open-label phase 2 study to evaluate UX007 in pediatric and adult patients with severe long-chain fatty acid oxidation disorders (LC-FAOD). *Journal of Inherited Metabolic Disease*, **2019**, 42, 169-177	5.4	26
175	A novel approach to the characterization of substrate specificity in short/branched chain Acyl-CoA dehydrogenase. *Journal of Biological Chemistry*, **2003**, 278, 37974-86	5.4	26

174	Purification and characterization of two polymorphic variants of short chain acyl-CoA dehydrogenase reveal reduction of catalytic activity and stability of the Gly185Ser enzyme. *Biochemistry*, **2002**, 41, 11126-33	3.2	26
173	Defect in dimethylglycine dehydrogenase, a new inborn error of metabolism: NMR spectroscopy study. *Clinical Chemistry*, **1999**, 45, 459-64	5.5	25
172	Safety and efficacy of omaveloxolone in patients with mitochondrial myopathy: MOTOR trial. *Neurology*, **2020**, 94, e687-e698	6.5	24
171	Different spectrum of mutations of isovaleryl-CoA dehydrogenase (IVD) gene in Korean patients with isovaleric acidemia. *Molecular Genetics and Metabolism*, **2007**, 92, 71-7	3.7	24
170	Leaky beta-oxidation of a trans-fatty acid: incomplete beta-oxidation of elaidic acid is due to the accumulation of 5-trans-tetradecenoyl-CoA and its hydrolysis and conversion to 5-trans-tetradecenoylcarnitine in the matrix of rat mitochondria. *Journal of Biological Chemistry*, **2004**, 279, 52160-7	5.4	24
169	Mitochondrial energetics is impaired in very long-chain acyl-CoA dehydrogenase deficiency and can be rescued by treatment with mitochondria-targeted electron scavengers. *Human Molecular Genetics*, **2019**, 28, 928-941	5.6	24
168	Blood ammonia and glutamine as predictors of hyperammonemic crises in patients with urea cycle disorder. *Genetics in Medicine*, **2015**, 17, 561-8	8.1	23
167	Functional role of the active site glutamate-368 in rat short chain acyl-CoA dehydrogenase. *Biochemistry*, **1996**, 35, 15356-63	3.2	23
166	Purification of human adult and foetal intestinal alkaline phosphatases by monoclonal antibody immunoaffinity chromatography. *Biochemical Journal*, **1984**, 217, 535-41	3.8	23
165	Human long chain, very long chain and medium chain acyl-CoA dehydrogenases are specific for the S-enantiomer of 2- methylpentadecanoyl-CoA. *Lipids and Lipid Metabolism*, **1998**, 1390, 333-8		22
164	Proteomic analysis of hyperdynamic mouse hearts with enhanced sarcoplasmic reticulum calcium cycling. *FASEB Journal*, **2004**, 18, 1725-7	0.9	22
163	Cloning of a gene for an acyl-CoA dehydrogenase from Pisum sativum L. and purification and characterization of its product as an isovaleryl-CoA dehydrogenase. *Journal of Biological Chemistry*, **2000**, 275, 33738-43	5.4	22
162	Defining clinical subgroups and genotype-phenotype correlations in NBAS-associated disease across 110 patients. *Genetics in Medicine*, **2020**, 22, 610-621	8.1	22
161	Metformin inhibits Branched Chain Amino Acid (BCAA) derived ketoacidosis and promotes metabolic homeostasis in MSUD. *Scientific Reports*, **2016**, 6, 28775	4.9	22
160	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. *American Journal of Human Genetics*, **2019**, 104, 815-834	11	21
159	The role of sterol-C4-methyl oxidase in epidermal biology. *Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids*, **2014**, 1841, 331-5	5	21
158	Low expression of long-chain acyl-CoA dehydrogenase in human skeletal muscle. *Molecular Genetics and Metabolism*, **2010**, 100, 163-7	3.7	21
157	Sandrow syndrome of mirror hands and feet and facial abnormalities. *American Journal of Medical Genetics Part A*, **1993**, 46, 126-8		21

156	Molecular characterization of four different classes of mutations in the isovaleryl-CoA dehydrogenase gene responsible for isovaleric acidemia. *American Journal of Human Genetics*, **1991**, 49, 147-57	11	20
155	Alterations in c-Myc phenotypes resulting from dynamin-related protein 1 (Drp1)-mediated mitochondrial fission. *Cell Death and Disease*, **2013**, 4, e670	9.8	19
154	Hepatic lymphangiomatosis mimicking polycystic liver disease. *Mayo Clinic Proceedings*, **1998**, 73, 1188-92	7.4	19
153	Normal N-acetylglutamate concentration measured in liver from a new patient with N-acetylglutamate synthetase deficiency: physiologic and biochemical implications. *Biochemical Medicine and Metabolic Biology*, **1992**, 47, 38-46		19
152	Abnormal lipid processing but normal long-term repopulation potential of myc-/- hepatocytes. *Oncotarget*, **2016**, 7, 30379-95	3.3	19
151	Efficient and gentle siRNA delivery by magnetofection. *Biotechnic and Histochemistry*, **2011**, 86, 226-31	1.8	18
150	Structural analysis of human adult and fetal alkaline phosphatases by cyanogen bromide peptide mapping. *Proceedings of the National Academy of Sciences of the United States of America*, **1984**, 81, 6120-3	11.5	18
149	The variant human isovaleryl-CoA dehydrogenase gene responsible for type II isovaleric acidemia determines an RNA splicing error, leading to the deletion of the entire second coding exon and the production of a truncated precursor protein that interacts poorly with mitochondrial import receptors.. *Journal of Biological Chemistry*, **1992**, 267, 2494-2501	5.4	18
148	Phenylketonuria: Current Treatments and Future Developments. *Drugs*, **2019**, 79, 495-500	12.1	17
147	Evolving Trends in Liver Transplant for Metabolic Liver Disease in the United States. *Liver Transplantation*, **2019**, 25, 911-921	4.5	17
146	Long-term safety and efficacy of pegvaliase for the treatment of phenylketonuria in adults: combined phase 2 outcomes through PAL-003 extension study. *Orphanet Journal of Rare Diseases*, **2018**, 13, 108	4.2	17
145	Prevalence and mutation analysis of short/branched chain acyl-CoA dehydrogenase deficiency (SBCADD) detected on newborn screening in Wisconsin. *Molecular Genetics and Metabolism*, **2013**, 110, 111-5	3.7	17
144	Recurrent vomiting and ethylmalonic aciduria associated with rare mutations of the short-chain acyl-CoA dehydrogenase gene. *Journal of Inherited Metabolic Disease*, **2003**, 26, 37-42	5.4	17
143	The variant human isovaleryl-CoA dehydrogenase gene responsible for type II isovaleric acidemia determines an RNA splicing error, leading to the deletion of the entire second coding exon and the production of a truncated precursor protein that interacts poorly with mitochondrial import receptors. *Journal of Biological Chemistry*, **1992**, 267, 2494-2501	5.4	17
142	Mechanism-based inhibitor discrimination in the acyl-CoA dehydrogenases. *Biochemistry*, **1997**, 36, 7761-8	3.2	16
141	Structure and analysis of the human dimethylglycine dehydrogenase gene. *Molecular Genetics and Metabolism*, **2000**, 69, 181-7	3.7	16
140	Central pontine myelinolysis as a complication of partial ornithine carbamoyl transferase deficiency. *American Journal of Medical Genetics Part A*, **1995**, 60, 210-3		16
139	Differentiation of human adult and fetal intestinal alkaline phosphatases with monoclonal antibodies. *American Journal of Human Genetics*, **1984**, 36, 987-1000	11	16

138	A randomized crossover trial of elamipretide in adults with primary mitochondrial myopathy. *Journal of Cachexia, Sarcopenia and Muscle*, **2020**, 11, 909-918	10.3	15
137	Possible Phenylacetate Hepatotoxicity During 4-Phenylbutyrate Therapy of Byler Disease. *Journal of Pediatric Gastroenterology and Nutrition*, **2016**, 62, 424-8	2.8	15
136	Therapies in inborn errors of oxidative metabolism. *Trends in Endocrinology and Metabolism*, **2012**, 23, 488-95	8.8	15
135	Systemic correction of a fatty acid oxidation defect by intramuscular injection of a recombinant adeno-associated virus vector. *Human Gene Therapy*, **2006**, 17, 71-80	4.8	15
134	Thermal unfolding of medium-chain acyl-CoA dehydrogenase and iso(3)valeryl-CoA dehydrogenase: study of the effect of genetic defects on enzyme stability. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2004**, 1690, 22-32	6.9	15
133	Short-chain hydroxyacyl-coenzyme A dehydrogenase deficiency presenting as unexpected infant death: A family study. *Journal of Pediatrics*, **2000**, 137, 257-9	3.6	15
132	Very long chain fatty acid metabolism is required in acute myeloid leukemia. *Blood*, **2021**, 137, 3518-3532	2.2	15
131	Safety and pharmacodynamics of an engineered E. coli Nissle for the treatment of phenylketonuria: a first-in-human phase 1/2a study. *Nature Metabolism*, **2021**, 3, 1125-1132	14.6	15
130	ETHE1 and MOCS1 deficiencies: Disruption of mitochondrial bioenergetics, dynamics, redox homeostasis and endoplasmic reticulum-mitochondria crosstalk in patient fibroblasts. *Scientific Reports*, **2019**, 9, 12651	4.9	14
129	Expanding research to provide an evidence base for nutritional interventions for the management of inborn errors of metabolism. *Molecular Genetics and Metabolism*, **2013**, 109, 319-28	3.7	14
128	Novel ETF dehydrogenase mutations in a patient with mild glutaric aciduria type II and complex II-III deficiency in liver and muscle. *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S481-7	5.4	14
127	Role of isovaleryl-CoA dehydrogenase and short branched-chain acyl-CoA dehydrogenase in the metabolism of valproic acid: implications for the branched-chain amino acid oxidation pathway. *Drug Metabolism and Disposition*, **2011**, 39, 1155-60	4	14
126	Identification of the catalytic residue of human short/branched chain acyl-CoA dehydrogenase by in vitro mutagenesis. *BBA - Proteins and Proteomics*, **1998**, 1382, 137-42		14
125	Investigating the link of ACAD10 deficiency to type 2 diabetes mellitus. *Journal of Inherited Metabolic Disease*, **2018**, 41, 49-57	5.4	13
124	Outcomes of cases with 3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency - Report from the Inborn Errors of Metabolism Information System. *Molecular Genetics and Metabolism*, **2016**, 118, 15-20	3.7	13
123	Unique plasma metabolomic signatures of individuals with inherited disorders of long-chain fatty acid oxidation. *Journal of Inherited Metabolic Disease*, **2016**, 39, 399-408	5.4	13
122	Altered DNA methylation in PAH deficient phenylketonuria. *Molecular Genetics and Metabolism*, **2015**, 115, 72-7	3.7	13
121	Potential misdiagnosis of 3-methylcrotonyl-coenzyme A carboxylase deficiency associated with absent or trace urinary 3-methylcrotonylglycine. *Pediatrics*, **2007**, 120, e1335-40	7.4	13

120	A porcine model of phenylketonuria generated by CRISPR/Cas9 genome editing. *JCI Insight*, **2020**, 5,	9.9	13
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