Rikke K Olsen

List of Publications by Citations

29 papers	1,198 citations	17 h-index	29 g-index
29 ext. papers	1,386 ext. citations	4.7 avg, IF	4.1 L-index

#	Paper	IF	Citations
29	ETFDH mutations as a major cause of riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. *Brain*, **2007**, 130, 2045-54	11.2	238
28	Clear relationship between ETF/ETFDH genotype and phenotype in patients with multiple acyl-CoA dehydrogenation deficiency. *Human Mutation*, **2003**, 22, 12-23	4.7	167
27	Mitochondrial fatty acid oxidation defects--remaining challenges. *Journal of Inherited Metabolic Disease*, **2008**, 31, 643-57	5.4	107
26	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. *American Journal of Human Genetics*, **2016**, 98, 1130-1145	11	97
25	Maternal riboflavin deficiency, resulting in transient neonatal-onset glutaric aciduria Type 2, is caused by a microdeletion in the riboflavin transporter gene GPR172B. *Human Mutation*, **2011**, 32, E1976-84	4.7	85
24	Molecular mechanisms of riboflavin responsiveness in patients with ETF-QO variations and multiple acyl-CoA dehydrogenation deficiency. *Human Molecular Genetics*, **2012**, 21, 3435-48	5.6	64
23	Role of flavinylation in a mild variant of multiple acyl-CoA dehydrogenation deficiency: a molecular rationale for the effects of riboflavin supplementation. *Journal of Biological Chemistry*, **2009**, 284, 4222-9	5.4	57
22	Redox signalling and mitochondrial stress responses; lessons from inborn errors of metabolism. *Journal of Inherited Metabolic Disease*, **2015**, 38, 703-19	5.4	51
21	Secondary coenzyme Q10 deficiency and oxidative stress in cultured fibroblasts from patients with riboflavin responsive multiple Acyl-CoA dehydrogenation deficiency. *Human Molecular Genetics*, **2013**, 22, 3819-27	5.6	48
20	Genetic and cellular modifiers of oxidative stress: what can we learn from fatty acid oxidation defects?. *Molecular Genetics and Metabolism*, **2013**, 110 Suppl, S31-9	3.7	42
19	An intronic variation in SLC52A1 causes exon skipping and transient riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. *Molecular Genetics and Metabolism*, **2017**, 122, 182-188	3.7	30
18	Short-chain acyl-CoA dehydrogenase deficiency: from gene to cell pathology and possible disease mechanisms. *Journal of Inherited Metabolic Disease*, **2017**, 40, 641-655	5.4	28
17	Riboflavin Deficiency-Implications for General Human Health and Inborn Errors of Metabolism. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	28
16	Cellular consequences of oxidative stress in riboflavin responsive multiple acyl-CoA dehydrogenation deficiency patient fibroblasts. *Human Molecular Genetics*, **2014**, 23, 4285-301	5.6	26
15	The ETFDH c.158A>G variation disrupts the balanced interplay of ESE- and ESS-binding proteins thereby causing missplicing and multiple Acyl-CoA dehydrogenation deficiency. *Human Mutation*, **2014**, 35, 86-95	4.7	22
14	Proteomic investigation of cultivated fibroblasts from patients with mitochondrial short-chain acyl-CoA dehydrogenase deficiency. *Molecular Genetics and Metabolism*, **2014**, 111, 360-368	3.7	19
13	Mitochondrial Spare Respiratory Capacity Is Negatively Correlated with Nuclear Reprogramming Efficiency. *Stem Cells and Development*, **2017**, 26, 166-176	4.4	18

List of Publications

12	Electron transfer flavoprotein and its role in mitochondrial energy metabolism in health and disease. *Gene*, **2021**, 776, 145407	3.8	13
11	FLAD1-associated multiple acyl-CoA dehydrogenase deficiency identified by newborn screening. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e915	2.3	11
10	Post-mortem detection of FLAD1 mutations in 2 Turkish siblings with hypotonia in early infancy. *Neuromuscular Disorders*, **2018**, 28, 787-790	2.9	11
9	DNA-based prenatal diagnosis for severe and variant forms of multiple acyl-CoA dehydrogenation deficiency. *Prenatal Diagnosis*, **2005**, 25, 60-4	3.2	9
8	High-resolution melting analysis, a simple and effective method for reliable mutation scanning and frequency studies in the ACADVL gene. *Journal of Inherited Metabolic Disease*, **2010**, 33, 247-60	5.4	8
7	FLAD1, encoding FAD synthase, is mutated in a patient with myopathy, scoliosis and cataracts. *Clinical Genetics*, **2018**, 94, 592-593	4	7
6	Bioenergetic and Proteomic Profiling of Immune Cells in Myalgic Encephalomyelitis/Chronic Fatigue Syndrome Patients: An Exploratory Study. *Biomolecules*, **2021**, 11,	5.9	3
5	Use of Molecular Genetic Analyses in Danish Routine Newborn Screening. *International Journal of Neonatal Screening*, **2021**, 7,	2.6	3
4	Increased antioxidant response in medium-chain acyl-CoA dehydrogenase deficiency: does lipoic acid have a protective role?. *Pediatric Research*, **2020**, 88, 556-564	3.2	2
3	Clinical, pathological and genetic features and follow-up of 110 patients with late-onset MADD: A single-center retrospective study. *Human Molecular Genetics*, **2021**,	5.6	2
2	Variants in the ethylmalonyl-CoA decarboxylase (ECHDC1) gene: a novel player in ethylmalonic aciduria?. *Journal of Inherited Metabolic Disease*, **2021**, 44, 1215-1225	5.4	2
1	Bezafibrate activation of PPAR drives disturbances in mitochondrial redox bioenergetics and decreases the viability of cells from patients with VLCAD deficiency. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2021**, 1867, 166100	6.9	0