

Rikke K Olsen

List of Publications by Year in descending order

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Version: 2024-02-01

29
papers

1,569
citations

430874

18
h-index

477307

29
g-index

29
all docs

29
docs citations

29
times ranked

1646
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical, pathological and genetic features and follow-up of 110 patients with late-onset MADD: a single-center retrospective study. <i>Human Molecular Genetics</i> , 2022, 31, 1115-1129.	2.9	10
2	Electron transfer flavoprotein and its role in mitochondrial energy metabolism in health and disease. <i>Gene</i> , 2021, 776, 145407.	2.2	42
3	Bezafibrate activation of PPAR drives disturbances in mitochondrial redox bioenergetics and decreases the viability of cells from patients with VLCAD deficiency. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2021, 1867, 166100.	3.8	5
4	Bioenergetic and Proteomic Profiling of Immune Cells in Myalgic Encephalomyelitis/Chronic Fatigue Syndrome Patients: An Exploratory Study. <i>Biomolecules</i> , 2021, 11, 961.	4.0	13
5	Variants in the <i>ethylmalonyl-CoA decarboxylase (ECHDC1)</i> gene: a novel player in ethylmalonic aciduria?. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1215-1225.	3.6	4
6	Use of Molecular Genetic Analyses in Danish Routine Newborn Screening. <i>International Journal of Neonatal Screening</i> , 2021, 7, 50.	3.2	12
7	Riboflavin Deficiency—Implications for General Human Health and Inborn Errors of Metabolism. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3847.	4.1	92
8	Increased antioxidant response in medium-chain acyl-CoA dehydrogenase deficiency: does lipoic acid have a protective role?. <i>Pediatric Research</i> , 2020, 88, 556-564.	2.3	4
9	<i>FLAD1</i> —associated multiple acyl-CoA dehydrogenase deficiency identified by newborn screening. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e915.	1.2	18
10	<i>FLAD1</i> , encoding FAD synthase, is mutated in a patient with myopathy, scoliosis and cataracts. <i>Clinical Genetics</i> , 2018, 94, 592-593.	2.0	9
11	Post-mortem detection of <i>FLAD1</i> mutations in 2 Turkish siblings with hypotonia in early infancy. <i>Neuromuscular Disorders</i> , 2018, 28, 787-790.	0.6	14
12	Short-chain acyl-CoA dehydrogenase deficiency: from gene to cell pathology and possible disease mechanisms. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 641-655.	3.6	41
13	An intronic variation in <i>SLC52A1</i> causes exon skipping and transient riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 182-188.	1.1	41
14	Mitochondrial Spare Respiratory Capacity Is Negatively Correlated with Nuclear Reprogramming Efficiency. <i>Stem Cells and Development</i> , 2017, 26, 166-176.	2.1	21
15	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. <i>American Journal of Human Genetics</i> , 2016, 98, 1130-1145.	6.2	118
16	Redox signalling and mitochondrial stress responses; lessons from inborn errors of metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 703-719.	3.6	59
17	The <i>ETFDH</i> c.158A>G Variation Disrupts the Balanced Interplay of ESE- and ESS-Binding Proteins thereby Causing Missplicing and Multiple Acyl-CoA Dehydrogenation Deficiency. <i>Human Mutation</i> , 2014, 35, 86-95.	2.5	32
18	Proteomic investigation of cultivated fibroblasts from patients with mitochondrial short-chain acyl-CoA dehydrogenase deficiency. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 360-368.	1.1	24

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19	Cellular consequences of oxidative stress in riboflavin responsive multiple acyl-CoA dehydrogenation deficiency patient fibroblasts. <i>Human Molecular Genetics</i> , 2014, 23, 4285-4301.	2.9	30
20	Genetic and cellular modifiers of oxidative stress: What can we learn from fatty acid oxidation defects?. <i>Molecular Genetics and Metabolism</i> , 2013, 110, S31-S39.	1.1	47
21	Secondary coenzyme Q10 deficiency and oxidative stress in cultured fibroblasts from patients with riboflavin responsive multiple Acyl-CoA dehydrogenation deficiency. <i>Human Molecular Genetics</i> , 2013, 22, 3819-3827.	2.9	59
22	Molecular mechanisms of riboflavin responsiveness in patients with ETF-QO variations and multiple acyl-CoA dehydrogenation deficiency. <i>Human Molecular Genetics</i> , 2012, 21, 3435-3448.	2.9	80
23	Maternal riboflavin deficiency, resulting in transient neonatal-onset glutaric aciduria Type 2, is caused by a microdeletion in the riboflavin transporter gene GPR172B. <i>Human Mutation</i> , 2011, 32, E1976-E1984.	2.5	96
24	High-resolution melting analysis, a simple and effective method for reliable mutation scanning and frequency studies in the <i>ACADVL</i> gene. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 247-260.	3.6	10
25	Role of Flavinylation in a Mild Variant of Multiple Acyl-CoA Dehydrogenation Deficiency. <i>Journal of Biological Chemistry</i> , 2009, 284, 4222-4229.	3.4	67
26	Mitochondrial fatty acid oxidation defects – remaining challenges. <i>Journal of Inherited Metabolic Disease</i> , 2008, 31, 643-657.	3.6	123
27	ETFDH mutations as a major cause of riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. <i>Brain</i> , 2007, 130, 2045-2054.	7.6	292
28	DNA-based prenatal diagnosis for severe and variant forms of multiple acyl-CoA dehydrogenation deficiency. <i>Prenatal Diagnosis</i> , 2005, 25, 60-64.	2.3	10
29	Clear relationship between ETF/ETFDH genotype and phenotype in patients with multiple acyl-CoA dehydrogenation deficiency. <i>Human Mutation</i> , 2003, 22, 12-23.	2.5	196