

Lisa G Riley

List of Publications by Year in descending order

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Version: 2024-02-01

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516710

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#	ARTICLE	IF	CITATIONS
1	Biallelic <i>AOPEP</i> Loss of Function Variants Cause Progressive Dystonia with Prominent Limb Involvement. <i>Movement Disorders</i> , 2022, 37, 137-147.	3.9	14
2	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. <i>Genetics in Medicine</i> , 2022, 24, 130-145.	2.4	45
3	FGF21 outperforms GDF15 as a diagnostic biomarker of mitochondrial disease in children. <i>Molecular Genetics and Metabolism</i> , 2022, 135, 63-71.	1.1	12
4	Deleterious variants in <i>CRLS1</i> lead to cardiolipin deficiency and cause an autosomal recessive multi-system mitochondrial disease. <i>Human Molecular Genetics</i> , 2022, 31, 3597-3612.	2.9	11
5	A novel <i>RLIM/RNF12</i> variant disrupts protein stability and function to cause severe Tonneau-Kalscheuer syndrome. <i>Scientific Reports</i> , 2021, 11, 9560.	3.3	5
6	Pre-clinical Investigation of Rett Syndrome Using Human Stem Cell-Based Disease Models. <i>Frontiers in Neuroscience</i> , 2021, 15, 698812.	2.8	10
7	A case of <i>QARS1</i> associated epileptic encephalopathy and review of epilepsy in aminoacyl-tRNA synthetase disorders. <i>Brain and Development</i> , 2021, , .	1.1	3
8	The diagnostic utility of genome sequencing in a pediatric cohort with suspected mitochondrial disease. <i>Genetics in Medicine</i> , 2020, 22, 1254-1261.	2.4	59
9	The expanding <i>LARS2</i> phenotypic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy. <i>Human Mutation</i> , 2020, 41, 1425-1434.	2.5	15
10	Biparental inheritance of mitochondrial DNA in humans is not a common phenomenon. <i>Genetics in Medicine</i> , 2019, 21, 2823-2826.	2.4	44
11	Biallelic variants in <i>LARS2</i> and <i>KARS</i> cause deafness and (ovario)leukodystrophy. <i>Neurology</i> , 2019, 92, e1225-e1237.	1.1	32
12	Clinical Spectrum and Functional Consequences Associated with Bi-Allelic Pathogenic <i>PNPT1</i> Variants. <i>Journal of Clinical Medicine</i> , 2019, 8, 2020.	2.4	16
13	Cryptic intronic <i>NBAS</i> variant reveals the genetic basis of recurrent liver failure in a child. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 77-82.	1.1	11
14	Tread carefully: A functional variant in the human NADPH oxidase 4 (<i>NOX4</i>) is not disease causing. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 382-387.	1.1	0
15	The phenotypic spectrum of germline <i>YARS2</i> variants: from isolated sideroblastic anemia to mitochondrial myopathy, lactic acidosis and sideroblastic anemia 2. <i>Haematologica</i> , 2018, 103, 2008-2015.	3.5	19
16	Squalene Synthase Deficiency: Clinical, Biochemical, and Molecular Characterization of a Defect in Cholesterol Biosynthesis. <i>American Journal of Human Genetics</i> , 2018, 103, 125-130.	6.2	29
17	A novel mutation in <i>GMPPA</i> in siblings with apparent intellectual disability, epilepsy, dysmorphism, and autonomic dysfunction. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2246-2250.	1.2	9
18	A <i>SLC39A8</i> variant causes manganese deficiency, and glycosylation and mitochondrial disorders. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 261-269.	3.6	101

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19	Progressive deafness“dystonia due to <i>SERAC1</i> mutations: A study of 67 cases. <i>Annals of Neurology</i> , 2017, 82, 1004-1015.	5.3	63
20	EPG5-Related Vici Syndrome: A Primary Defect of Autophagic Regulation with an Emerging Phenotype Overlapping with Mitochondrial Disorders. <i>JIMD Reports</i> , 2017, 42, 19-29.	1.5	7
21	Compound heterozygous mutations in glycyI-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. <i>PLoS ONE</i> , 2017, 12, e0178125.	2.5	16
22	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. <i>American Journal of Human Genetics</i> , 2016, 99, 1229-1244.	6.2	91
23	Whole Exome Sequencing Identifies the Genetic Basis of Late-Onset Leigh Syndrome in a Patient with MRI but Little Biochemical Evidence of a Mitochondrial Disorder. <i>JIMD Reports</i> , 2016, 32, 117-124.	1.5	11
24	Delayed diagnosis of congenital myasthenia due to associated mitochondrial enzyme defect. <i>Neuromuscular Disorders</i> , 2015, 25, 257-261.	0.6	16
25	Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. <i>Human Molecular Genetics</i> , 2015, 24, 2297-2307.	2.9	64
26	LARS2 Variants Associated with Hydrops, Lactic Acidosis, Sideroblastic Anemia, and Multisystem Failure. <i>JIMD Reports</i> , 2015, 28, 49-57.	1.5	48
27	Rapid Identification of a Novel Complex I MT-ND3 m.10134C>A Mutation in a Leigh Syndrome Patient. <i>PLoS ONE</i> , 2014, 9, e104879.	2.5	5
28	Mitochondrial respiratory chain disorders in childhood: Insights into diagnosis and management in the new era of genomic medicine. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2014, 1840, 1368-1379.	2.4	34
29	Mutations in CYC1, Encoding Cytochrome c1 Subunit of Respiratory Chain Complex III, Cause Insulin-Responsive Hyperglycemia. <i>American Journal of Human Genetics</i> , 2013, 93, 384-389.	6.2	61
30	Mutations in LYRM4, encoding iron“sulfur cluster biogenesis factor ISD11, cause deficiency of multiple respiratory chain complexes. <i>Human Molecular Genetics</i> , 2013, 22, 4460-4473.	2.9	97
31	Phenotypic variability and identification of novel YARS2 mutations in YARS2 mitochondrial myopathy, lactic acidosis and sideroblastic anaemia. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 193.	2.7	49
32	Mutation of the Mitochondrial Tyrosyl-tRNA Synthetase Gene, YARS2, Causes Myopathy, Lactic Acidosis, and Sideroblastic Anemia“MLASA Syndrome. <i>American Journal of Human Genetics</i> , 2010, 87, 52-59.	6.2	211