Lisa G Riley

List of Publications by Year
in descending order

32 papers	1,208 citations	516710 16 h-index	434195 31 g-index
32 all docs	32 docs citations	32 times ranked	2223 citing authors

#	Article	IF	Citations
1	Mutation of the Mitochondrial Tyrosyl-tRNA Synthetase Gene, YARS2, Causes Myopathy, Lactic Acidosis, and Sideroblastic Anemiaâ€"MLASA Syndrome. American Journal of Human Genetics, 2010, 87, 52-59.	6.2	211
2	A SLC39A8 variant causes manganese deficiency, and glycosylation and mitochondrial disorders. Journal of Inherited Metabolic Disease, 2017, 40, 261-269.	3.6	101
3	Mutations in LYRM4, encoding ironâ€"sulfur cluster biogenesis factor ISD11, cause deficiency of multiple respiratory chain complexes. Human Molecular Genetics, 2013, 22, 4460-4473.	2.9	97
4	MECR Mutations Cause Childhood-Onset Dystonia and Optic Atrophy, a Mitochondrial Fatty Acid Synthesis Disorder. American Journal of Human Genetics, 2016, 99, 1229-1244.	6.2	91
5	Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. Human Molecular Genetics, 2015, 24, 2297-2307.	2.9	64
6	Progressive deafnessâ€"dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
7	Mutations in CYC1, Encoding Cytochrome c1 Subunit of Respiratory Chain Complex III, Cause Insulin-Responsive Hyperglycemia. American Journal of Human Genetics, 2013, 93, 384-389.	6.2	61
8	The diagnostic utility of genome sequencing in a pediatric cohort with suspected mitochondrial disease. Genetics in Medicine, 2020, 22, 1254-1261.	2.4	59
9	Phenotypic variability and identification of novel YARS2 mutations in YARS2 mitochondrial myopathy, lactic acidosis and sideroblastic anaemia. Orphanet Journal of Rare Diseases, 2013, 8, 193.	2.7	49
10	LARS2 Variants Associated with Hydrops, Lactic Acidosis, Sideroblastic Anemia, and Multisystem Failure. JIMD Reports, 2015, 28, 49-57.	1.5	48
11	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	2.4	45
12	Biparental inheritance of mitochondrial DNA in humans is not a common phenomenon. Genetics in Medicine, 2019, 21, 2823-2826.	2.4	44
13	Mitochondrial respiratory chain disorders in childhood: Insights into diagnosis and management in the new era of genomic medicine. Biochimica Et Biophysica Acta - General Subjects, 2014, 1840, 1368-1379.	2.4	34
14	Biallelic variants in LARS2 and KARS cause deafness and (ovario)leukodystrophy. Neurology, 2019, 92, e1225-e1237.	1.1	32
15	Squalene Synthase Deficiency: Clinical, Biochemical, and Molecular Characterization of a Defect in Cholesterol Biosynthesis. American Journal of Human Genetics, 2018, 103, 125-130.	6.2	29
16	The phenotypic spectrum of germline YARS2 variants: from isolated sideroblastic anemia to mitochondrial myopathy, lactic acidosis and sideroblastic anemia 2. Haematologica, 2018, 103, 2008-2015.	3.5	19
17	Delayed diagnosis of congenital myasthenia due to associated mitochondrial enzyme defect. Neuromuscular Disorders, 2015, 25, 257-261.	0.6	16
18	Clinical Spectrum and Functional Consequences Associated with Bi-Allelic Pathogenic PNPT1 Variants. Journal of Clinical Medicine, 2019, 8, 2020.	2.4	16

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19	Compound heterozygous mutations in glycyl-tRNA synthetase (GARS) cause mitochondrial respiratory chain dysfunction. PLoS ONE, 2017, 12, e0178125.	2.5	16
20	The expandingLARS2phenotypic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy. Human Mutation, 2020, 41, 1425-1434.	2.5	15
21	Biallelic <scp>AOPEP</scp> Lossâ€ofâ€Function Variants Cause Progressive Dystonia with Prominent Limb Involvement. Movement Disorders, 2022, 37, 137-147.	3.9	14
22	FGF21 outperforms GDF15 as a diagnostic biomarker of mitochondrial disease in children. Molecular Genetics and Metabolism, 2022, 135, 63-71.	1.1	12
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. JIMD Reports, 2016, 32, 117-124.	1.5	11
24	Cryptic intronic NBAS variant reveals the genetic basis of recurrent liver failure in a child. Molecular Genetics and Metabolism, 2019, 126, 77-82.	1.1	11
25	Deleterious variants in CRLS1 lead to cardiolipin deficiency and cause an autosomal recessive multi-system mitochondrial disease. Human Molecular Genetics, 2022, 31, 3597-3612.	2.9	11
26	Pre-clinical Investigation of Rett Syndrome Using Human Stem Cell-Based Disease Models. Frontiers in Neuroscience, 2021, 15, 698812.	2.8	10
27	A novel mutation in GMPPA in siblings with apparent intellectual disability, epilepsy, dysmorphism, and autonomic dysfunction. American Journal of Medical Genetics, Part A, 2017, 173, 2246-2250.	1.2	9
28	EPG5-Related Vici Syndrome: A Primary Defect of Autophagic Regulation with an Emerging Phenotype Overlapping with Mitochondrial Disorders. JIMD Reports, 2017, 42, 19-29.	1.5	7
29	Rapid Identification of a Novel Complex I MT-ND3 m.10134C>A Mutation in a Leigh Syndrome Patient. PLoS ONE, 2014, 9, e104879.	2.5	5
30	A novel RLIM/RNF12 variant disrupts protein stability and function to cause severe Tonneâ€“Kalscheuer syndrome. Scientific Reports, 2021, 11, 9560.	3.3	5
31	A case of QARS1 associated epileptic encephalopathy and review of epilepsy in aminoacyl-tRNA synthetase disorders. Brain and Development, 2021, , .	1.1	3
32	Tread carefully: A functional variant in the human NADPH oxidase 4 (NOX4) is not disease causing. Molecular Genetics and Metabolism, 2018, 123, 382-387.	1.1	0