Lindsay C Burrage

List of Publications by Year
in descending order

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71	2,970	186265 28	197818 49
papers	citations	h-index	g-index

79	79	79	6068
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
2	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. Human Molecular Genetics, 2014, 23, R1-R8.	2.9	234
3	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181
4	Untargeted metabolomic analysis for the clinical screening of inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2015, 38, 1029-1039.	3.6	169
5	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
6	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	10.2	108
7	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583.	6.2	92
8	A recurrent p.Arg92Trp variant in steroidogenic factor-1 (NR5A1) can act as a molecular switch in human sex development. Human Molecular Genetics, 2016, 25, 3446-3453.	2.9	90
9	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	8.2	87
10	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	8.1	71
11	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913.	6.2	65
12	Recurrent ACADVL molecular findings in individuals with a positive newborn screen for very long chain acyl-coA dehydrogenase (VLCAD) deficiency in the United States. Molecular Genetics and Metabolism, 2015, 116, 139-145.	1.1	65
13	Dominant-negative mutations in human IL6ST underlie hyper-IgE syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	64
14	Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia (MLASA) plus associated with a novel de novo mutation (m.8969G>A) in the mitochondrial encoded ATP6 gene. Molecular Genetics and Metabolism, 2014, 113, 207-212.	1.1	63
15	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
16	High prevalence of overweight and obesity in females with phenylketonuria. Molecular Genetics and Metabolism, 2012, 107, 43-48.	1.1	58
17	Sodium phenylbutyrate decreases plasma branched-chain amino acids in patients with urea cycle disorders. Molecular Genetics and Metabolism, 2014, 113, 131-135.	1.1	58
18	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	6.2	55

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19	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	6.2	54
20	Inherited Metabolic Disorders. Nutrition in Clinical Practice, 2015, 30, 502-510.	2.4	53
21	Untargeted metabolomic profiling reveals multiple pathway perturbations and new clinical biomarkers in urea cycle disorders. Genetics in Medicine, 2019, 21, 1977-1986.	2.4	47
22	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	8.2	43
23	Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. Human Molecular Genetics, 2015, 24, 6417-6427.	2.9	40
24	Argininosuccinate Lyase Deficiency Causes an Endothelial-Dependent Form of Hypertension. American Journal of Human Genetics, 2018, 103, 276-287.	6.2	39
25	Congenital hyperinsulinism as the presenting feature of Kabuki syndrome: clinical and molecular characterization of 10 affected individuals. Genetics in Medicine, 2019, 21, 233-242.	2.4	39
26	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. Journal of Clinical Investigation, 2021, 131, .	8.2	39
27	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
28	Liver transplantation in propionic and methylmalonic acidemia: A single center study with literature review. Molecular Genetics and Metabolism, 2019, 128, 431-443.	1.1	36
29	Lung Transplantation for FLNA -Associated Progressive Lung Disease. Journal of Pediatrics, 2017, 186, 118-123.e6.	1.8	32
30	Elevations of C14:1 and C14:2 Plasma Acylcarnitines in Fasted Children: AÂDiagnostic Dilemma. Journal of Pediatrics, 2016, 169, 208-213.e2.	1.8	30
31	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	2.5	27
32	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
33	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	8.2	23
34	De Novo Variants in CDK19 Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. American Journal of Human Genetics, 2020, 106, 717-725.	6.2	23
35	Lysinuric protein intolerance presenting with multiple fractures. Molecular Genetics and Metabolism Reports, 2014, 1, 176-183.	1.1	20
36	Milder clinical and biochemical phenotypes associated with the c.482G > A (p.Arg161Gln) pathogenic variant in cobalamin C disease: Implications for management and screening. Molecular Genetics and Metabolism, 2017, 122, 60-66.	1.1	20

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37	Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in MT-ATP6. Mitochondrion, 2019, 44, 58-64.	3.4	19
38	Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2018, 103, 1030-1037.	6.2	18
39	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	6.2	18
40	A mosaic 2q24.2 deletion narrows the critical region to a 0.4â€‰Mb interval that includes TBR1, TANK, and PSMD14. American Journal of Medical Genetics, Part A, 2013, 161, 841-844.	1.2	17
41	Neonatal fractures as a presenting feature of LMOD3â€associated congenital myopathy. American Journal of Medical Genetics, Part A, 2017, 173, 2789-2794.	1.2	17
42	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. Molecular Genetics & Genomic Medicine, 2020, 8, e1397.	1.2	16
43	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	2.4	16
44	A global Slc7a7 knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. Human Molecular Genetics, 2020, 29, 2171-2184.	2.9	15
45	Clinical diagnosis of metabolic disorders using untargeted metabolomic profiling and disease-specific networks learned from profiling data. Scientific Reports, 2022, 12, 6556.	3.3	15
46	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
47	Immune Dysregulation Mimicking Systemic Lupus Erythematosus in a Patient With Lysinuric Protein Intolerance: Case Report and Review of the Literature. Frontiers in Pediatrics, 2021, 9, 673957.	1.9	12
48	De novo variants in CACNA1E found in patients with intellectual disability, developmental regression and social cognition deficit but no seizures. Molecular Autism, 2021, 12, 69.	4.9	12
49	Nineteen-year follow-up of a patient with severe glutathione synthetase deficiency. Journal of Human Genetics, 2016, 61, 669-672.	2.3	10
50	Chronic liver disease and impaired hepatic glycogen metabolism in argininosuccinate lyase deficiency. JCI Insight, 2020, 5, .	5.0	10
51	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	1.2	9
52	A dominant negative variant of RAB5B disrupts maturation of surfactant protein B and surfactant protein C. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	9
53	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. American Journal of Medical Genetics, Part A, 2017, 173, 733-739.	1.2	8
54	Microdeletions excluding YWHAE and PAFAH1B1 cause a unique leukoencephalopathy: further delineation of the 17p13.3 microdeletion spectrum. Genetics in Medicine, 2019, 21, 1652-1656.	2.4	8

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55	A deep intronic variant is a common cause of OTC deficiency in individuals with previously negative genetic testing. Molecular Genetics and Metabolism Reports, 2021, 26, 100706.	1.1	8
56	Biomarkers for liver disease in urea cycle disorders. Molecular Genetics and Metabolism, 2021, 133, 148-156.	1.1	8
57	Catelâ€“Manzke syndrome: Further delineation of the phenotype associated with pathogenic variants in TGDS. Molecular Genetics and Metabolism Reports, 2015, 4, 89-91.	1.1	7
58	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
59	Adult presentation of Xâ€linked Conradiâ€HÃ¼nermannâ€Happle syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1309-1314.	1.2	6
60	Novel deletion of 6p21.31p21.1 associated with laryngeal cleft, developmental delay, dysmorphic features and vascular anomaly. European Journal of Medical Genetics, 2019, 62, 103531.	1.3	4
61	GNA11 brain somatic pathogenic variant in an individual with phacomatosis pigmentovascularis. Neurology: Genetics, 2019, 5, e366.	1.9	4
62	Multisite Retrospective Review of Outcomes in Renal Replacement Therapy for Neonates with Inborn Errors of Metabolism. Journal of Pediatrics, 2022, 246, 116-122.e1.	1.8	4
63	Functional analysis of a novel de novo variant in PPP5C associated with microcephaly, seizures, and developmental delay. Molecular Genetics and Metabolism, 2022, 136, 65-73.	1.1	4
64	A novel, de novo intronic variant in <scp>POGZ</scp> causes <scp>Whiteâ€“Sutton</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2198-2203.	1.2	4
65	De novo interstitial duplication of 15q11.2â€“q13.1 with complex maternal uniparental trisomy for the 15q11â€“q13 region in a patient with Praderâ€–Willi syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2557-2563.	1.2	2
66	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2315-2324.	1.2	2
67	Application of lung volume reduction surgery for a child with filamin A (FLNA) mutations. Pediatric Pulmonology, 2022, 57, 224-230.	2.0	2
68	PRUNE1 c.<scp>933G</scp>>A synonymous variant induces exon 7 skipping, disrupts the <scp>DHHA2</scp> domain, and leads to an atypical <scp>NMIHBA</scp> syndrome presentation: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 1868-1874.	1.2	2
69	Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation. Journal of Genetic Counseling, 2022, 31, 326-337.	1.6	1
70	Maffucci Syndrome, Calcium Homeostasis, and Endocrine Challenges in Management. Journal of the Endocrine Society, 2021, 5, A701-A701.	0.2	0
71	SAT-LB088 Assessing Metacarpal Cortical Thickness as a Tool to Evaluate Bone Density Compared to DXA in Osteogenesis Imperfecta. Journal of the Endocrine Society, 2019, 3, .	0.2	0