Lindsay C Burrage

List of Publications by Year
in descending order

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	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
2	Application of lung volume reduction surgery for a child with filamin A (FLNA) mutations. Pediatric Pulmonology, 2022, 57, 224-230.	2.0	2
3	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
4	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
5	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
6	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
7	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
8	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
9	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	8.2	87
10	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
11	Nitric oxide modulates bone anabolism through regulation of osteoblast glycolysis and differentiation. Journal of Clinical Investigation, 2021, 131, .	8.2	39
12	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	1.2	9
13	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
14	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
15	Maffucci Syndrome, Calcium Homeostasis, and Endocrine Challenges in Management. Journal of the Endocrine Society, 2021, 5, A701-A701.	0.2	0
16	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
17	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	2.4	16
18	Biomarkers for liver disease in urea cycle disorders. Molecular Genetics and Metabolism, 2021, 133, 148-156.	1.1	8

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19	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	6.2	18
20	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
21	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
22	Dominant-negative mutations in human IL6ST underlie hyper-IgE syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	64
23	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
24	A global Slc7a7 knockout mouse model demonstrates characteristic phenotypes of human lysinuric protein intolerance. Human Molecular Genetics, 2020, 29, 2171-2184.	2.9	15
25	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
26	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	8.1	71
27	Chronic liver disease and impaired hepatic glycogen metabolism in argininosuccinate lyase deficiency. JCI Insight, 2020, 5, .	5.0	10
28	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
29	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
30	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
31	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
32	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
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	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
35	GNA11 brain somatic pathogenic variant in an individual with phacomatosis pigmentovascularis. Neurology: Genetics, 2019, 5, e366.	1.9	4
36	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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37	Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in MT-ATP6. Mitochondrion, 2019, 44, 58-64.	3.4	19
38	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
39	Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2018, 103, 1030-1037.	6.2	18
40	Argininosuccinate Lyase Deficiency Causes an Endothelial-Dependent Form of Hypertension. American Journal of Human Genetics, 2018, 103, 276-287.	6.2	39
41	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
42	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
43	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
44	Lung Transplantation for FLNA -Associated Progressive Lung Disease. Journal of Pediatrics, 2017, 186, 118-123.e6.	1.8	32
45	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
46	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
47	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
48	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
49	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	2.5	27
50	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. American Journal of Medical Genetics, Part A, 2017, 173, 733-739.	1.2	8
51	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
52	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	8.2	43
53	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
54	Nineteen-year follow-up of a patient with severe glutathione synthetase deficiency. Journal of Human Genetics, 2016, 61, 669-672.	2.3	10

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55	Adult presentation of Xâ€linked Conradiâ€HÃ¼nermannâ€Happle syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1309-1314.	1.2	6
56	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
57	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
58	Inherited Metabolic Disorders. Nutrition in Clinical Practice, 2015, 30, 502-510.	2.4	53
59	Untargeted metabolomic analysis for the clinical screening of inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2015, 38, 1029-1039.	3.6	169
60	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	6.2	55
61	Human recombinant arginase enzyme reduces plasma arginine in mouse models of arginase deficiency. Human Molecular Genetics, 2015, 24, 6417-6427.	2.9	40
62	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
63	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	10.2	108
64	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. Human Molecular Genetics, 2014, 23, R1-R8.	2.9	234
65	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
66	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
67	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
68	Lysinuric protein intolerance presenting with multiple fractures. Molecular Genetics and Metabolism Reports, 2014, 1, 176-183.	1.1	20
69	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
70	High prevalence of overweight and obesity in females with phenylketonuria. Molecular Genetics and Metabolism, 2012, 107, 43-48.	1.1	58
71	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