Lynne A Wolfe

List of Publications by Year
in descending order

66	4,024	156536	150775
papers	citations	32 h-index	59 g-index

68	68	68	8410
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Novel CUL3 Variant Causing Familial Hyperkalemic Hypertension Impairs Regulation and Function of Ubiquitin Ligase Activity. Hypertension, 2022, 79, 60-75.	1.3	17
2	eP198: EIF3F compound heterozygous genotype-phenotype association. Genetics in Medicine, 2022, 24, S123.	1.1	1
3	Diagnosis and discovery: Insights from the <scp>NIH</scp> Undiagnosed Diseases Program. Journal of Inherited Metabolic Disease, 2022, 45, 907-918.	1.7	2
4	An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. Genetics in Medicine, 2021, 23, 740-750.	1.1	25
5	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	1.1	18
6	Compound heterozygous KCTD7 variants in progressive myoclonus epilepsy. Journal of Neurogenetics, 2021, 35, 74-83.	0.6	4
7	FOXR1 regulates stress response pathways and is necessary for proper brain development. PLoS Genetics, 2021, 17, e1009854.	1.5	3
8	Predominant and novel de novo variants in 29 individuals with <scp>ALG13</scp> deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. Journal of Inherited Metabolic Disease, 2020, 43, 1333-1348.	1.7	24
9	Mutations in GET4 disrupt the transmembrane domain recognition complex pathway. Journal of Inherited Metabolic Disease, 2020, 43, 1037-1045.	1.7	4
10	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
11	Cell-based analysis of CAD variants identifies individuals likely to benefit from uridine therapy. Genetics in Medicine, 2020, 22, 1598-1605.	1.1	18
12	Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. Molecular Genetics and Metabolism, 2020, 130, 49-57.	0.5	15
13	Defining the clinical phenotype of Saulâ€“Wilson syndrome. Genetics in Medicine, 2020, 22, 857-866.	1.1	11
14	Deficiency in the endocytic adaptor proteins PHETA1/2 impair renal and craniofacial development. DMM Disease Models and Mechanisms, 2020, 13, .	1.2	7
15	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	1.1	60
16	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	0.8	6
17	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	1.1	19
18	IgG4â€related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686.	0.6	8

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19	Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy. PLoS ONE, 2019, 14, e0214250.	1.1	59
20	Arrest of Fetal Brain Development in ALG11-Congenital Disorder of Glycosylation. Pediatric Neurology, 2019, 94, 64-69.	1.0	3
21	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	1.1	39
22	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
23	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	2.6	59
24	Early infantile-onset epileptic encephalopathy 28 due to a homozygous microdeletion involving the WWOX gene in a region of uniparental disomy. Human Mutation, 2019, 40, 42-47.	1.1	8
25	PARP1 inhibition alleviates injury in ARH3-deficient mice and human cells. JCI Insight, 2019, 4, .	2.3	34
26	Defective glycosylation and multisystem abnormalities characterize the primary immunodeficiency XMEN disease. Journal of Clinical Investigation, 2019, 130, 507-522.	3.9	74
27	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	2.6	59
28	Urine oligosaccharide screening by MALDI-TOF for the identification of NGLY1 deficiency. Molecular Genetics and Metabolism, 2018, 124, 82-86.	0.5	29
29	A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. American Journal of Human Genetics, 2018, 103, 553-567.	2.6	58
30	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	2.6	56
31	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	2.6	69
32	A recurrent de novo missense mutation in UBTF causes developmental neuroregression. Human Molecular Genetics, 2018, 27, 691-705.	1.4	32
33	Prospective phenotyping of NGLY1-CDDG, the first congenital disorder of deglycosylation. Genetics in Medicine, 2017, 19, 160-168.	1.1	124
34	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	2.6	142
35	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	2.6	35
36	De novo missense variants inHECW2are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86.	1.5	46

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37	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.	2.6	181
38	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	2.6	96
39	Defining Disease, Diagnosis, and Translational Medicine within a Homeostatic Perturbation Paradigm: The National Institutes of Health Undiagnosed Diseases Program Experience. Frontiers in Medicine, 2017, 4, 62.	1.2	23
40	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	1.1	40
41	Nutritional interventions in primary mitochondrial disorders: Developing an evidence base. Molecular Genetics and Metabolism, 2016, 119, 187-206.	0.5	41
42	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	2.6	68
43	Aberrant splicing induced by the most commonEPG5mutation in an individual with Vici syndrome. Brain, 2016, 139, e52-e52.	3.7	14
44	Mitotic Intragenic Recombination: A Mechanism of Survival for Several Congenital Disorders of Glycosylation. American Journal of Human Genetics, 2016, 98, 339-346.	2.6	14
45	Disruption of Golgi morphology and altered protein glycosylation in PLA2G6-associated neurodegeneration. Journal of Medical Genetics, 2016, 53, 180-189.	1.5	27
46	Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. Genetics in Medicine, 2016, 18, 608-617.	1.1	85
47	Biallelic mutations in CAD, impair de novo pyrimidine biosynthesis and decrease glycosylation precursors. Human Molecular Genetics, 2015, 24, 3050-3057.	1.4	66
48	Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2015, 17, 689-701.	1.1	414
49	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	0.5	76
50	Expanding the clinical and molecular characteristics of PIGT-CDG, a disorder of glycosylphosphatidylinositol anchors. Molecular Genetics and Metabolism, 2015, 115, 128-140.	0.5	44
51	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	2.6	61
52	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. Nature Genetics, 2014, 46, 503-509.	9.4	490
53	Practice patterns of mitochondrial disease physicians in North America. Part 1: Diagnostic and clinical challenges. Mitochondrion, 2014, 14, 26-33.	1.6	36
54	Autosomal recessive phosphoglucomutase 3 (PGM3) mutations link glycosylation defects to atopy, immune deficiency, autoimmunity, and neurocognitive impairment. Journal of Allergy and Clinical Immunology, 2014, 133, 1400-1409.e5.	1.5	193

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55	Glycosylation, Hypogammaglobulinemia, and Resistance to Viral Infections. New England Journal of Medicine, 2014, 370, 1615-1625.	13.9	117
56	The implications of familial incidental findings from exome sequencing: the NIH Undiagnosed Diseases Program experience. Genetics in Medicine, 2014, 16, 741-750.	1.1	56
57	Three rare diseases in one Sib pair: RAI1, PCK1, GRIN2B mutations associated with Smithâ€“Magenis Syndrome, cytosolic PEPCK deficiency and NMDA receptor glutamate insensitivity. Molecular Genetics and Metabolism, 2014, 113, 161-170.	0.5	58
58	Two novel compound heterozygous mutations in OPA3 in two siblings with OPA3-related 3-methylglutaconic aciduria. Molecular Genetics and Metabolism Reports, 2014, 1, 114-123.	0.4	6
59	The Elusive Magic Pill: Finding Effective Therapies for Mitochondrial Disorders. Neurotherapeutics, 2013, 10, 320-328.	2.1	13
60	Practice patterns of mitochondrial disease physicians in North America. Part 2: treatment, care and management. Mitochondrion, 2013, 13, 681-687.	1.6	38
61	Congenital disorders of glycosylation and intellectual disability. Developmental Disabilities Research Reviews, 2013, 17, 211-225.	2.9	145
62	Novel SNP array analysis and exome sequencing detect a homozygous exon 7 deletion of MEGF10 causing early onset myopathy, areflexia, respiratory distress and dysphagia (EMARDD). Neuromuscular Disorders, 2013, 23, 483-488.	0.3	22
63	The National Institutes of Health Undiagnosed Diseases Program: insights into rare diseases. Genetics in Medicine, 2012, 14, 51-59.	1.1	254
64	LCR-initiated rearrangements at the IDS locus, completed with Alu-mediated recombination or non-homologous end joining. Journal of Human Genetics, 2011, 56, 516-523.	1.1	10
65	Mutations in the human SC4MOL gene encoding a methyl sterol oxidase cause psoriasiform dermatitis, microcephaly, and developmental delay. Journal of Clinical Investigation, 2011, 121, 976-984.	3.9	91
66	Potential Misdiagnosis of 3-Methylcrotonyl-Coenzyme A Carboxylase Deficiency Associated With Absent or Trace Urinary 3-Methylcrotonylglycine. Pediatrics, 2007, 120, e1335-e1340.	1.0	17