

Hane Lee

List of Publications by Year in descending order

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Version: 2024-02-01

98
papers

8,383
citations

66234

42
h-index

48187

88
g-index

103
all docs

103
docs citations

103
times ranked

16777
citing authors

#	ARTICLE	IF	CITATIONS
1	Melanomas acquire resistance to B-RAF(V600E) inhibition by RTK or N-RAS upregulation. <i>Nature</i> , 2010, 468, 973-977.	13.7	1,944
2	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. <i>JAMA - Journal of the American Medical Association</i> , 2014, 312, 1880.	3.8	842
3	Cold Urticaria, Immunodeficiency, and Autoimmunity Related to <i>PLCG2</i> Deletions. <i>New England Journal of Medicine</i> , 2012, 366, 330-338.	13.9	391
4	U87MG Decoded: The Genomic Sequence of a Cytogenetically Aberrant Human Cancer Cell Line. <i>PLoS Genetics</i> , 2010, 6, e1000832.	1.5	229
5	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. <i>Nature Genetics</i> , 2012, 44, 575-580.	9.4	212
6	Mutations in <i>PYCR1</i> cause cutis laxa with progeroid features. <i>Nature Genetics</i> , 2009, 41, 1016-1021.	9.4	211
7	Exome Sequencing in the Clinical Diagnosis of Sporadic or Familial Cerebellar Ataxia. <i>JAMA Neurology</i> , 2014, 71, 1237.	4.5	211
8	SGK196 Is a Glycosylation-Specific <i>O</i> -Mannose Kinase Required for Dystroglycan Function. <i>Science</i> , 2013, 341, 896-899.	6.0	197
9	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. <i>American Journal of Human Genetics</i> , 2017, 100, 843-853.	2.6	181
10	Exome Sequencing for the Diagnosis of 46,XY Disorders of Sex Development. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E333-E344.	1.8	172
11	Mutations in the PCNA-binding domain of <i>CDKN1C</i> cause IMAGE syndrome. <i>Nature Genetics</i> , 2012, 44, 788-792.	9.4	169
12	Multiple self-healing squamous epithelioma is caused by a disease-specific spectrum of mutations in <i>TGFBR1</i> . <i>Nature Genetics</i> , 2011, 43, 365-369.	9.4	147
13	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 185-192.	2.6	142
14	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. <i>Genetics in Medicine</i> , 2020, 22, 490-499.	1.1	136
15	Exome Sequencing Identifies <i>PDE4D</i> Mutations in Acrodysostosis. <i>American Journal of Human Genetics</i> , 2012, 90, 746-751.	2.6	128
16	Assessing the necessity of confirmatory testing for exome-sequencing results in a clinical molecular diagnostic laboratory. <i>Genetics in Medicine</i> , 2014, 16, 510-515.	1.1	121
17	Accuracy of phenotyping of autistic children based on internet implemented parent report. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 1119-1126.	1.1	120
18	Disruption of <i>POF1B</i> Binding to Nonmuscle Actin Filaments Is Associated with Premature Ovarian Failure. <i>American Journal of Human Genetics</i> , 2006, 79, 113-119.	2.6	116

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19	De Novo Nonsense Mutations in KAT6A, a Lysine Acetyl-Transferase Gene, Cause a Syndrome Including Microcephaly and Global Developmental Delay. <i>American Journal of Human Genetics</i> , 2015, 96, 498-506.	2.6	115
20	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. <i>European Journal of Human Genetics</i> , 2015, 23, 1473-1481.	1.4	101
21	The functional O-mannose glycan on Î±-dystroglycan contains a phospho-ribitol primed for matriglycan addition. <i>ELife</i> , 2016, 5, .	2.8	98
22	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. <i>American Journal of Human Genetics</i> , 2017, 100, 128-137.	2.6	96
23	Mutations in TFAM, encoding mitochondrial transcription factor A, cause neonatal liver failure associated with mtDNA depletion. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 91-99.	0.5	93
24	Next-generation mapping: a novel approach for detection of pathogenic structural variants with a potential utility in clinical diagnosis. <i>Genome Medicine</i> , 2017, 9, 90.	3.6	86
25	Loss of CHSY1, a Secreted FRINGE Enzyme, Causes Syndromic Brachydactyly in Humans via Increased NOTCH Signaling. <i>American Journal of Human Genetics</i> , 2010, 87, 768-778.	2.6	82
26	Sequence variant in the laminin Î³1 (LAMC1) gene associated with familial pelvic organ prolapse. <i>Human Genetics</i> , 2007, 120, 847-856.	1.8	76
27	<i>EXTL3</i> mutations cause skeletal dysplasia, immune deficiency, and developmental delay. <i>Journal of Experimental Medicine</i> , 2017, 214, 623-637.	4.2	76
28	Clinical application of next-generation sequencing to the practice of neurology. <i>Lancet Neurology</i> , The, 2019, 18, 492-503.	4.9	76
29	IRF2BPL Is Associated with Neurological Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 245-260.	2.6	69
30	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. <i>Nature Genetics</i> , 2012, 44, 709-713.	9.4	68
31	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 991-999.	2.6	68
32	A New Episodic Ataxia Syndrome With Linkage to Chromosome 19q13. <i>Archives of Neurology</i> , 2007, 64, 749.	4.9	65
33	Expanding the phenotype of mutations in DICER1: mosaic missense mutations in the RNase IIIb domain of <i>DICER1</i> cause GLOW syndrome. <i>Journal of Medical Genetics</i> , 2014, 51, 294-302.	1.5	65
34	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. <i>Genetics in Medicine</i> , 2019, 21, 161-172.	1.1	60
35	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 494-504.	2.6	59
36	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. <i>American Journal of Human Genetics</i> , 2019, 104, 1127-1138.	2.6	59

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37	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	2.6	59
38	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. <i>Human Mutation</i> , 2020, 41, 487-501.	1.1	58
39	<i>KMT2B</i> -related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. <i>Brain</i> , 2020, 143, 3242-3261.	3.7	57
40	A genome-wide linkage scan of familial benign recurrent vertigo: linkage to 22q12 with evidence of heterogeneity. <i>Human Molecular Genetics</i> , 2006, 15, 251-258.	1.4	56
41	A homozygous loss-of-function CAMK2A mutation causes growth delay, frequent seizures and severe intellectual disability. <i>ELife</i> , 2018, 7, .	2.8	53
42	Scaling resolution of variant classification differences in ClinVar between 41 clinical laboratories through an outlier approach. <i>Human Mutation</i> , 2018, 39, 1641-1649.	1.1	50
43	A novel mutation in KCNA1 causes episodic ataxia without myokymia. <i>Human Mutation</i> , 2004, 24, 536-536.	1.1	47
44	Phenotypic and Genetic Analysis of a Large Family With Migraine-Associated Vertigo. <i>Headache</i> , 2008, 48, 1460-1467.	1.8	46
45	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. <i>American Journal of Human Genetics</i> , 2019, 105, 413-424.	2.6	43
46	Missense-depleted regions in population exomes implicate ras superfamily nucleotide-binding protein alteration in patients with brain malformation. <i>Npj Genomic Medicine</i> , 2016, 1, .	1.7	41
47	SLC35A2-CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. <i>Human Mutation</i> , 2019, 40, 908-925.	1.1	39
48	Effects of a Mutation in the HSPE1 Gene Encoding the Mitochondrial Co-chaperonin HSP10 and Its Potential Association with a Neurological and Developmental Disorder. <i>Frontiers in Molecular Biosciences</i> , 2016, 3, 65.	1.6	38
49	The PTH/PTHrP-SIK3 pathway affects skeletogenesis through altered mTOR signaling. <i>Science Translational Medicine</i> , 2018, 10, .	5.8	38
50	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. <i>Cilia</i> , 2016, 5, 8.	1.8	37
51	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. <i>American Journal of Human Genetics</i> , 2020, 106, 570-583.	2.6	37
52	Disseminated Coccidioidomycosis Treated with Interferon- β and Dupilumab. <i>New England Journal of Medicine</i> , 2020, 382, 2337-2343.	13.9	36
53	Association of progesterone receptor with migraine-associated vertigo. <i>Neurogenetics</i> , 2007, 8, 195-200.	0.7	35
54	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. <i>American Journal of Human Genetics</i> , 2017, 100, 343-351.	2.6	35

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55	Improving the efficiency of genomic loci capture using oligonucleotide arrays for high throughput resequencing. <i>BMC Genomics</i> , 2009, 10, 646.	1.2	34
56	Loss of the scavenger mRNA decapping enzyme DCPS causes syndromic intellectual disability with neuromuscular defects. <i>Human Molecular Genetics</i> , 2015, 24, 3163-3171.	1.4	31
57	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. <i>American Journal of Human Genetics</i> , 2019, 105, 854-868.	2.6	29
58	Variant in human POFUT1 reduces enzymatic activity and likely causes a recessive microcephaly, global developmental delay with cardiac and vascular features. <i>Glycobiology</i> , 2018, 28, 276-283.	1.3	24
59	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. <i>Annals of the New York Academy of Sciences</i> , 2016, 1366, 49-60.	1.8	23
60	Estimating the relative frequency of leukodystrophies and recommendations for carrier screening in the era of next-generation sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1906-1912.	0.7	22
61	GATAD2B-associated neurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-related disorder. <i>Genetics in Medicine</i> , 2020, 22, 878-888.	1.1	22
62	Heterozygous variants in <i>MYBPC1</i> are associated with an expanded neuromuscular phenotype beyond arthrogryposis. <i>Human Mutation</i> , 2019, 40, 1115-1126.	1.1	19
63	Truncating mutations in <i>APP</i> cause a distinct neurological phenotype. <i>Annals of Neurology</i> , 2016, 80, 456-460.	2.8	18
64	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2018, 103, 948-967.	2.6	18
65	Whole genome sequencing reveals novel <i>IGHMBP2</i> variant leading to unique cryptic splice site and Charcot-Marie-Tooth phenotype with early onset symptoms. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00676.	0.6	18
66	De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. <i>American Journal of Human Genetics</i> , 2021, 108, 1330-1341.	2.6	18
67	Pathogenic variants in <i>SMARCA5</i> , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. <i>Science Advances</i> , 2021, 7, .	4.7	17
68	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. <i>Genetics in Medicine</i> , 2021, 23, 1075-1085.	1.1	16
69	Pierpont syndrome associated with the p.Tyr446Cys missense mutation in TBL1XR1. <i>European Journal of Medical Genetics</i> , 2017, 60, 504-508.	0.7	15
70	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. <i>American Journal of Human Genetics</i> , 2020, 107, 544-554.	2.6	13
71	Next generation sequencing in clinical diagnosis. <i>Lancet Neurology</i> , The, 2019, 18, 426.	4.9	11
72	Detection of a mosaic <i>CDKL5</i> deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1665.	0.6	11

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73	Mitchell-Riley syndrome iPSC exhibit reduced pancreatic endoderm differentiation due to an <i>RFX6</i> mutation. <i>Development (Cambridge)</i> , 2020, 147, .	1.2	10
74	A second locus for schneckenbecken dysplasia identified by a mutation in the gene encoding inositol polyphosphate phosphatase-like 1 (<i>INPPL1</i>). <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2470-2473.	0.7	9
75	Early Infantile Epileptic Encephalopathy with a de novo variant in ZEB2 identified by exome sequencing. <i>European Journal of Medical Genetics</i> , 2016, 59, 70-74.	0.7	8
76	Congenital myasthenic syndrome caused by a frameshift insertion mutation in <i>GFPT1</i> . <i>Neurology: Genetics</i> , 2020, 6, e468.	0.9	8
77	<i>DYRK1A</i> pathogenic variants in two patients with syndromic intellectual disability and a review of the literature. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1544.	0.6	8
78	Expansion of <i>NEUROD2</i> phenotypes to include developmental delay without seizures. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1076-1080.	0.7	7
79	Segmental overgrowth and aneurysms due to mosaic <i>PDGFRB</i> p.(Tyr562Cys). <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1430-1436.	0.7	7
80	The frontiers of sequencing in undiagnosed neurodevelopmental diseases. <i>Current Opinion in Genetics and Development</i> , 2020, 65, 76-83.	1.5	6
81	Myopathy associated with homozygous <i>PYROXD1</i> pathogenic variants detected by genome sequencing. <i>Neuropathology</i> , 2020, 40, 302-307.	0.7	6
82	Novel <i>NUDT2</i> variant causes intellectual disability and polyneuropathy. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 2320-2325.	1.7	5
83	<i>MPEG1/Perforin-2</i> Haploinsufficiency Associated Polymicrobial Skin Infections and Considerations for Interferon- β Therapy. <i>Frontiers in Immunology</i> , 2020, 11, 601584.	2.2	5
84	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 136.	1.2	5
85	Rethinking clinical practice: clinical implementation of exome sequencing. <i>Personalized Medicine</i> , 2012, 9, 785-787.	0.8	4
86	Genetic characterization and long-term management of severely affected siblings with intellectual developmental disorder with cardiac arrhythmia syndrome. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 23, 100582.	0.4	4
87	Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. <i>Journal of Genetic Counseling</i> , 2021, 30, 439-447.	0.9	4
88	Mutation in TWINKLE in a Large Iranian Family with Progressive External Ophthalmoplegia, Myopathy, Dysphagia and Dysphonia, and Behavior Change. <i>Archives of Iranian Medicine</i> , 2016, 19, 87-91.	0.2	4
89	An infant with <i>MLH1</i> variants, <i>FOXG1</i> duplication and multiple, benign cranial and spinal tumors: A clinical exome sequencing study. <i>Genes Chromosomes and Cancer</i> , 2016, 55, 131-142.	1.5	3
90	Leveraging ancestry to improve causal variant identification in exome sequencing for monogenic disorders. <i>European Journal of Human Genetics</i> , 2016, 24, 113-119.	1.4	3

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91	Novel association of familial testicular germ cell tumor and autosomal dominant polycystic kidney disease with <i>PKD1</i> mutation. <i>Pediatric Blood and Cancer</i> , 2017, 64, 100-102.	0.8	3
92	Maternal Uniparental Disomy 14 (UPD14) Identified by Clinical Exome Sequencing in an Adolescent with Diverticulosis. <i>ACG Case Reports Journal</i> , 2019, 6, e00021.	0.2	3
93	Biallelic alterations in <i>PLXND1</i> cause common arterial trunk and other cardiac malformations in humans. <i>Human Molecular Genetics</i> , 2023, 32, 353-356.	1.4	3
94	A homozygous in-frame duplication within the LRRCT consensus sequence of <i>CFAP410</i> causes cone-rod dystrophy, macular staphyloma and short stature. <i>Ophthalmic Genetics</i> , 2022, 43, 378-384.	0.5	3
95	OPTICAL COHERENCE TOMOGRAPHY AND OPTICAL COHERENCE TOMOGRAPHY ANGIOGRAPHY FINDINGS AND VISUAL PROGNOSIS IN TWO PATIENTS WITH POSTERIOR MICROPHthalmOS. <i>Retinal Cases and Brief Reports</i> , 2022, 16, 253-257.	0.3	2
96	A Sodium Channel Myotonia Presenting with Intermittent Dysphagia as a Manifestation of a Rare SCN4A Variant. <i>Journal of Molecular Neuroscience</i> , 2017, 61, 312-314.	1.1	1
97	Case Report: Whole Exome Sequencing Identifies Compound Heterozygous Variants in TSM Gene Causing Juvenile Hypertrophic Cardiomyopathy. <i>Frontiers in Cardiovascular Medicine</i> , 2021, 8, 798985.	1.1	1
98	RABENOSYN separation-of-function mutations uncouple endosomal recycling from lysosomal degradation, causing a distinct Mendelian Disorder. <i>Human Molecular Genetics</i> , 0, , .	1.4	0