Hane Lee

List of Publications by Year
in descending order

98 papers	8,383 citations	66343 42 h-index	48315 88 g-index
103 all docs	103 docs citations	103 times ranked	16777 citing authors

#	Article	IF	Citations
1	Biallelic alterations in PLXND1 cause common arterial trunk and other cardiac malformations in humans. Human Molecular Genetics, 2023, 32, 353-356.	2.9	3
2	OPTICAL COHERENCE TOMOGRAPHY AND OPTICAL COHERENCE TOMOGRAPHY ANGIOGRAPHY FINDINGS AND VISUAL PROGNOSIS IN TWO PATIENTS WITH POSTERIOR MICROPHTHALMOS. Retinal Cases and Brief Reports, 2022, 16, 253-257.	0.6	2
3	A homozygous in-frame duplication within the LRRCT consensus sequence of CFAP410 causes cone-rod dystrophy, macular staphyloma and short stature. Ophthalmic Genetics, 2022, 43, 378-384.	1.2	3
4	Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. Journal of Genetic Counseling, 2021, 30, 439-447.	1.6	4
5	Expansion of <scp>NEUROD2</scp> phenotypes to include developmental delay without seizures. American Journal of Medical Genetics, Part A, 2021, 185, 1076-1080.	1.2	7
6	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine, 2021, 23, 1075-1085.	2.4	16
7	Segmental overgrowth and aneurysms due to mosaic PDGFRB p.(Tyr562Cys). American Journal of Medical Genetics, Part A, 2021, 185, 1430-1436.	1.2	7
8	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136.	2.7	5
9	Pathogenic variants in SMARCA5 , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, .	10.3	17
10	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	1.2	11
11	De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 1330-1341.	6.2	18
12	Case Report: Whole Exome Sequencing Identifies Compound Heterozygous Variants in TSFM Gene Causing Juvenile Hypertrophic Cardiomyopathy. Frontiers in Cardiovascular Medicine, 2021, 8, 798985.	2.4	1
13	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. Genetics in Medicine, 2020, 22, 490-499.	2.4	136
14	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	2.5	58
15	Congenital myasthenic syndrome caused by a frameshift insertion mutation in GFPT1. Neurology: Genetics, 2020, 6, e468.	1.9	8
16	Mitchell-Riley syndrome iPSC exhibit reduced pancreatic endoderm differentiation due to an RFX6 mutation. Development (Cambridge), 2020, 147, .	2.5	10
17	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. American Journal of Human Genetics, 2020, 107, 544-554.	6.2	13
18	Novel NUDT2 variant causes intellectual disability and polyneuropathy. Annals of Clinical and Translational Neurology, 2020, 7, 2320-2325.	3.7	5

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19	MPEG1/Perforin-2 Haploinsufficiency Associated Polymicrobial Skin Infections and Considerations for Interferon-Î³ Therapy. Frontiers in Immunology, 2020, 11, 601584.	4.8	5
20	DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature. Molecular Genetics & Genomic Medicine, 2020, 8, e1544.	1.2	8
21	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
22	Genetic characterization and long-term management of severely affected siblings with intellectual developmental disorder with cardiac arrhythmia syndrome. Molecular Genetics and Metabolism Reports, 2020, 23, 100582.	1.1	4
23	Estimating the relative frequency of leukodystrophies and recommendations for carrier screening in the era of nextâ€generation sequencing. American Journal of Medical Genetics, Part A, 2020, 182, 1906-1912.	1.2	22
24	Disseminated Coccidioidomycosis Treated with Interferon-Î³ and Dupilumab. New England Journal of Medicine, 2020, 382, 2337-2343.	27.0	36
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	The frontiers of sequencing in undiagnosed neurodevelopmental diseases. Current Opinion in Genetics and Development, 2020, 65, 76-83.	3.3	6
27	Myopathy associated with homozygous PYROXD1 pathogenic variants detected by genome sequencing. Neuropathology, 2020, 40, 302-307.	1.2	6
28	GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888.	2.4	22
29	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
30	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
31	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	6.2	29
32	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	6.2	59
33	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	2.5	19
34	Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic spliceâ€site and Charcotâ€Marieâ€Tooth phenotype with early onset symptoms. Molecular Genetics & Genomic Medicine, 2019, 7, e00676.	1.2	18
35	Clinical application of next-generation sequencing to the practice of neurology. Lancet Neurology, The, 2019, 18, 492-503.	10.2	76
36	Next generation sequencing in clinical diagnosis. Lancet Neurology, The, 2019, 18, 426.	10.2	11

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37	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
38	Maternal Uniparental Disomy 14 (UPD14) Identified by Clinical Exome Sequencing in an Adolescent with Diverticulosis. ACG Case Reports Journal, 2019, 6, e00021.	0.4	3
39	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.	6.2	59
40	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
41	Variant in human POFUT1 reduces enzymatic activity and likely causes a recessive microcephaly, global developmental delay with cardiac and vascular features. Glycobiology, 2018, 28, 276-283.	2.5	24
42	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 948-967.	6.2	18
43	Scaling resolution of variant classification differences in ClinVar between 41 clinical laboratories through an outlier approach. Human Mutation, 2018, 39, 1641-1649.	2.5	50
44	The PTH/PTHrP-SIK3 pathway affects skeletogenesis through altered mTOR signaling. Science Translational Medicine, 2018, 10, .	12.4	38
45	A homozygous loss-of-function CAMK2A mutation causes growth delay, frequent seizures and severe intellectual disability. ELife, 2018, 7, .	6.0	53
46	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
47	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
48	EXTL3 mutations cause skeletal dysplasia, immune deficiency, and developmental delay. Journal of Experimental Medicine, 2017, 214, 623-637.	8.5	76
49	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.	6.2	35
50	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
51	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
52	A Sodium Channel Myotonia Presenting with Intermittent Dysphagia as a Manifestation of a Rare SCN4A Variant. Journal of Molecular Neuroscience, 2017, 61, 312-314.	2.3	1
53	Pierpont syndrome associated with the p.Tyr446Cys missense mutation in TBL1XR1. European Journal of Medical Genetics, 2017, 60, 504-508.	1.3	15
54	Novel association of familial testicular germ cell tumor and autosomal dominant polycystic kidney disease with PKD1 mutation. Pediatric Blood and Cancer, 2017, 64, 100-102.	1.5	3

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55	Next-generation mapping: a novel approach for detection of pathogenic structural variants with a potential utility in clinical diagnosis. Genome Medicine, 2017, 9, 90.	8.2	86
56	Effects of a Mutation in the HSPE1 Gene Encoding the Mitochondrial Co-chaperonin HSP10 and Its Potential Association with a Neurological and Developmental Disorder. Frontiers in Molecular Biosciences, 2016, 3, 65.	3.5	38
57	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. Annals of the New York Academy of Sciences, 2016, 1366, 49-60.	3.8	23
58	An infant with <scp>MLH</scp>3 variants, <scp>FOXG</scp>1â€duplication and multiple, benign cranial and spinal tumors: A clinical exome sequencing study. Genes Chromosomes and Cancer, 2016, 55, 131-142.	2.8	3
59	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	6.2	68
60	Truncating mutations in APP cause a distinct neurological phenotype. Annals of Neurology, 2016, 80, 456-460.	5.3	18
61	Mutations in TFAM, encoding mitochondrial transcription factor A, cause neonatal liver failure associated with mtDNA depletion. Molecular Genetics and Metabolism, 2016, 119, 91-99.	1.1	93
62	Missense-depleted regions in population exomes implicate ras superfamily nucleotide-binding protein alteration in patients with brain malformation. Npj Genomic Medicine, 2016, 1, .	3.8	41
63	A novel ICK mutation causes ciliary disruption and lethal endocrine-cerebro-osteodysplasia syndrome. Cilia, 2016, 5, 8.	1.8	37
64	Early Infantile Epileptic Encephalopathy with a de novo variant in ZEB2 identified by exome sequencing. European Journal of Medical Genetics, 2016, 59, 70-74.	1.3	8
65	Leveraging ancestry to improve causal variant identification in exome sequencing for monogenic disorders. European Journal of Human Genetics, 2016, 24, 113-119.	2.8	3
66	The functional O-mannose glycan on Î±-dystroglycan contains a phospho-ribitol primed for matriglycan addition. ELife, 2016, 5, .	6.0	98
67	Mutation in TWINKLE in a Large Iranian Family with Progressive External Ophthalmoplegia, Myopathy, Dysphagia and Dysphonia, and Behavior Change. Archives of Iranian Medicine, 2016, 19, 87-91.	0.6	4
68	A second locus for schneckenbecken dysplasia identified by a mutation in the gene encoding inositol polyphosphate phosphataseâ€like 1 (INPPL1). American Journal of Medical Genetics, Part A, 2015, 167, 2470-2473.	1.2	9
69	Loss of the scavenger mRNA decapping enzyme DCPS causes syndromic intellectual disability with neuromuscular defects. Human Molecular Genetics, 2015, 24, 3163-3171.	2.9	31
70	De Novo Nonsense Mutations in KAT6A, a Lysine Acetyl-Transferase Gene, Cause a Syndrome Including Microcephaly and Global Developmental Delay. American Journal of Human Genetics, 2015, 96, 498-506.	6.2	115
71	Exome Sequencing for the Diagnosis of 46,XY Disorders of Sex Development. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E333-E344.	3.6	172
72	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. European Journal of Human Genetics, 2015, 23, 1473-1481.	2.8	101

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73	Exome Sequencing in the Clinical Diagnosis of Sporadic or Familial Cerebellar Ataxia. JAMA Neurology, 2014, 71, 1237.	9.0	211
74	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	7.4	842
75	Assessing the necessity of confirmatory testing for exome-sequencing results in a clinical molecular diagnostic laboratory. Genetics in Medicine, 2014, 16, 510-515.	2.4	121
76	Expanding the phenotype of mutations in DICER1: mosaic missense mutations in the RNase IIIb domain ofDICER1cause GLOW syndrome. Journal of Medical Genetics, 2014, 51, 294-302.	3.2	65
77	SGK196 Is a Glycosylation-Specific O -Mannose Kinase Required for Dystroglycan Function. Science, 2013, 341, 896-899.	12.6	197
78	Mutations in the PCNA-binding domain of CDKN1C cause IMAGe syndrome. Nature Genetics, 2012, 44, 788-792.	21.4	169
79	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. Nature Genetics, 2012, 44, 575-580.	21.4	212
80	Rethinking clinical practice: clinical implementation of exome sequencing. Personalized Medicine, 2012, 9, 785-787.	1.5	4
81	Cold Urticaria, Immunodeficiency, and Autoimmunity Related toPLCG2Deletions. New England Journal of Medicine, 2012, 366, 330-338.	27.0	391
82	Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. Nature Genetics, 2012, 44, 709-713.	21.4	68
83	Exome Sequencing Identifies PDE4D Mutations in Acrodysostosis. American Journal of Human Genetics, 2012, 90, 746-751.	6.2	128
84	Multiple self-healing squamous epithelioma is caused by a disease-specific spectrum of mutations in TGFBR1. Nature Genetics, 2011, 43, 365-369.	21.4	147
85	Loss of CHSY1, a Secreted FRINGE Enzyme, Causes Syndromic Brachydactyly in Humans via Increased NOTCH Signaling. American Journal of Human Genetics, 2010, 87, 768-778.	6.2	82
86	Accuracy of phenotyping of autistic children based on internet implemented parent report. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 1119-1126.	1.7	120
87	Melanomas acquire resistance to B-RAF(V600E) inhibition by RTK or N-RAS upregulation. Nature, 2010, 468, 973-977.	27.8	1,944
88	U87MG Decoded: The Genomic Sequence of a Cytogenetically Aberrant Human Cancer Cell Line. PLoS Genetics, 2010, 6, e1000832.	3.5	229
89	Improving the efficiency of genomic loci capture using oligonucleotide arrays for high throughput resequencing. BMC Genomics, 2009, 10, 646.	2.8	34
90	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	21.4	211

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91	Phenotypic and Genetic Analysis of a Large Family With Migraineâ€Associated Vertigo. Headache, 2008, 48, 1460-1467.	3.9	46
92	A New Episodic Ataxia Syndrome With Linkage to Chromosome 19q13. Archives of Neurology, 2007, 64, 749.	4.5	65
93	Sequence variant in the laminin Î³1 (LAMC1) gene associated with familial pelvic organ prolapse. Human Genetics, 2007, 120, 847-856.	3.8	76
94	Association of progesterone receptor with migraine-associated vertigo. Neurogenetics, 2007, 8, 195-200.	1.4	35
95	Disruption of POF1B Binding to Nonmuscle Actin Filaments Is Associated with Premature Ovarian Failure. American Journal of Human Genetics, 2006, 79, 113-119.	6.2	116
96	A genome-wide linkage scan of familial benign recurrent vertigo: linkage to 22q12 with evidence of heterogeneity. Human Molecular Genetics, 2006, 15, 251-258.	2.9	56
97	A novel mutation in KCNA1 causes episodic ataxia without myokymia. Human Mutation, 2004, 24, 536-536.	2.5	47
98	RABENOSYN separation-of-function mutations uncouple endosomal recycling from lysosomal degradation, causing a distinct Mendelian Disorder. Human Molecular Genetics, 0, , .	2.9	0