Pengfei Liu

List of Publications by Year
in descending order

124 papers	8,798 citations	61857 43 h-index	48187 88 g-index
136 all docs	136 docs citations	136 times ranked	13784 citing authors

#	Article	IF	Citations
1	Clinical exome sequencing data reveal high diagnostic yields for congenital diaphragmatic hernia plus (CDH+) and new phenotypic expansions involving CDH. Journal of Medical Genetics, 2022, 59, 270-278.	1.5	27
2	Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples. Journal of Genetic Counseling, 2022, 31, 59-70.	0.9	3
3	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373.	1.1	12
4	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.0	14
5	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
6	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. Npj Genomic Medicine, 2022, 7, 11.	1.7	7
7	Genome sequencing reveals novel noncoding variants in <scp>PLA2G6</scp> and <scp>LMNB1</scp> causing progressive neurologic disease. Molecular Genetics & Genomic Medicine, 2022, 10, e1892.	0.6	4
8	Emerging technologies for prenatal diagnosis: The application of whole genome and RNA sequencing. Prenatal Diagnosis, 2022, 42, 686-696.	1.1	6
9	Biallelic variants in WARS1 cause a highly variable neurodevelopmental syndrome and implicate a critical exon for normal auditory function. Human Mutation, 2022, 43, 1472-1489.	1.1	6
10	Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). Journal of Medical Genetics, 2021, 58, 41-47.	1.5	40
11	Combined Genome Sequencing and RNA Analysis Reveals and Characterizes a Deep Intronic Variant in IGHMBP2 in a Patient With Spinal Muscular Atrophy With Respiratory Distress Type 1. Pediatric Neurology, 2021, 114, 16-20.	1.0	7
12	Amino Acid-Level Signal-to-Noise Analysis Aids in Pathogenicity Prediction of Incidentally Identified TTN -Encoded Titin Truncating Variants. Circulation Genomic and Precision Medicine, 2021, 14, e003131.	1.6	7
13	Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014.	1.0	10
14	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	1.1	18
15	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	3.9	87
16	Identification and characterization of novel ACD variants: modulation of TPP1 protein level offsets the impact of germline loss-of-function variants on telomere length. Journal of Physical Education and Sports Management, 2021, 7, a005454.	0.5	8
17	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine, 2021, 23, 1075-1085.	1.1	16
18	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30

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19	Perturbations of genes essential for MÃ¼llerian duct and WÃ¶lffian duct development in Mayer-Rokitansky-KÃ¼ster-Hauser syndrome. American Journal of Human Genetics, 2021, 108, 337-345.	2.6	41
20	Clinical characterization of individuals with the distal 1q21.1 microdeletion. American Journal of Medical Genetics, Part A, 2021, 185, 1388-1398.	0.7	6
21	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. Frontiers in Cell and Developmental Biology, 2021, 9, 631428.	1.8	4
22	Expanding the phenotype, genotype and biochemical knowledge of <scp>ALG3â€CDG</scp>. Journal of Inherited Metabolic Disease, 2021, 44, 987-1000.	1.7	4
23	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
24	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	0.7	9
25	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2315-2324.	0.7	2
26	Exome sequencing reveals genetic architecture in patients with isolated or syndromic short stature. Journal of Genetics and Genomics, 2021, 48, 396-402.	1.7	21
27	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	0.6	11
28	Saturation mutagenesis defines novel mouse models of severe spine deformity. DMM Disease Models and Mechanisms, 2021, 14, .	1.2	4
29	Contribution of uniparental disomy in a clinical trio exome cohort of 2675 patients. Molecular Genetics & Genomic Medicine, 2021, 9, e1792.	0.6	4
30	Vertical transmission of a large calvarial ossification defect due to heterozygous variants of ALX4 and TWIST1. American Journal of Medical Genetics, Part A, 2021, 185, 916-922.	0.7	1
31	Detection of low-level parental somatic mosaicism for clinically relevant SNVs and indels identified in a large exome sequencing dataset. Human Genomics, 2021, 15, 72.	1.4	11
32	De novo variants in H3-3A and H3-3B are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. Npj Genomic Medicine, 2021, 6, 104.	1.7	7
33	Exome sequencing reveals a novel variant in NFX1 causing intracranial aneurysm in a Chinese family. Journal of NeuroInterventional Surgery, 2020, 12, 221-226.	2.0	7
34	TBX6 missense variants expand the mutational spectrum in a nonâ€Mendelian inheritance disease. Human Mutation, 2020, 41, 182-195.	1.1	27
35	Bi-allelic Mutations in NADSYN1 Cause Multiple Organ Defects and Expand the Genotypic Spectrum of Congenital NAD Deficiency Disorders. American Journal of Human Genetics, 2020, 106, 129-136.	2.6	27
36	Front Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, i.	1.1	0

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37	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	3.7	33
38	Recurrent arginine substitutions in the ACTG2 gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654.	1.1	27
39	Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in FBN1. Molecular Genetics & Genomic Medicine, 2020, 8, e1023.	0.6	19
40	Analysis of the role of the human papillomavirus 16/18 E7 protein assay in screening for cervical intraepithelial neoplasia: a case control study. BMC Cancer, 2020, 20, 999.	1.1	7
41	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
42	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. Genetics in Medicine, 2020, 22, 1768-1776.	1.1	30
43	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. Npj Genomic Medicine, 2020, 5, 47.	1.7	67
44	Phenotypic expansion in KIF1A â€related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104.	1.1	8
45	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. Expert Review of Molecular Diagnostics, 2020, 20, 995-1002.	1.5	14
46	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.	2.6	23
47	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	1.1	36
48	Cell-based analysis of CAD variants identifies individuals likely to benefit from uridine therapy. Genetics in Medicine, 2020, 22, 1598-1605.	1.1	18
49	SGLT2 Inhibitors for Treatment of Refractory Hypomagnesemia: A Case Report of 3 Patients. Kidney Medicine, 2020, 2, 359-364.	1.0	29
50	Genome sequencing analysis of a family with a child displaying severe abdominal distention and recurrent hypoglycemia. Molecular Genetics & Genomic Medicine, 2020, 8, e1130.	0.6	5
51	Increased TBX6 gene dosages induce congenital cervical vertebral malformations in humans and mice. Journal of Medical Genetics, 2020, 57, 371-379.	1.5	23
52	Human and mouse studies establish TBX6 in Mendelian CAKUT and as a potential driver of kidney defects associated with the 16p11.2 microdeletion syndrome. Kidney International, 2020, 98, 1020-1030.	2.6	17
53	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
54	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	1.1	15

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55	Biallelic and De Novo Variants in DONSON Reveal a Clinical Spectrum of Cell Cycleâ€opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. American Journal of Medical Genetics, Part A, 2019, 179, 2056-2066.	0.7	15
56	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	3.6	55
57	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. American Journal of Human Genetics, 2019, 105, 384-394.	2.6	37
58	DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract. Genetics in Medicine, 2019, 21, 2755-2764.	1.1	19
59	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	2.6	30
60	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	4.7	35
61	Loss of function of NCOR1 and NCOR2 impairs memory through a novel GABAergic hypothalamusâ€“CA3 projection. Nature Neuroscience, 2019, 22, 205-217.	7.1	54
62	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
63	Review of the phenotypic spectrum associated with haploinsufficiency of MYRF. American Journal of Medical Genetics, Part A, 2019, 179, 1376-1382.	0.7	44
64	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	3.6	42
65	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.	3.6	23
66	De novo missense variant in the GTPase effector domain (GED) of DNM1L leads to static encephalopathy and seizures. Journal of Physical Education and Sports Management, 2019, 5, a003673.	0.5	24
67	Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. Cell, 2019, 176, 1310-1324.e10.	13.5	73
68	Validation Studies for Single Circulating Trophoblast Genetic Testing as a Form of Noninvasive Prenatal Diagnosis. American Journal of Human Genetics, 2019, 105, 1262-1273.	2.6	47
69	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	1.1	52
70	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
71	TBX6-associated congenital scoliosis (TACS) as a clinically distinguishable subtype of congenital scoliosis: further evidence supporting the compound inheritance and TBX6 gene dosage model. Genetics in Medicine, 2019, 21, 1548-1558.	1.1	60
72	Monozygotic twins with non-Down syndrome associated MLL-rearranged hematologic malignancy and megakaryoblastic differentiation. Leukemia and Lymphoma, 2019, 60, 1083-1086.	0.6	3

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73	Expanding the FANCO/RAD51C associated phenotype: Cleft lip and palate and lobar holoprosencephaly, two rare findings in Fanconi anemia. European Journal of Medical Genetics, 2018, 61, 257-261.	0.7	11
74	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
75	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74.	3.6	105
76	Villoglandular adenocarcinoma of cervix: pathologic features, clinical management, and outcome. Cancer Management and Research, 2018, Volume 10, 3955-3961.	0.9	10
77	Phenotypic expansion in <scp>DDX</scp>3X â€“ a common cause of intellectual disability in females. Annals of Clinical and Translational Neurology, 2018, 5, 1277-1285.	1.7	66
78	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
79	The coexistence of copy number variations (CNVs) and single nucleotide polymorphisms (SNPs) at a locus can result in distorted calculations of the significance in associating SNPs to disease. Human Genetics, 2018, 137, 553-567.	1.8	57
80	Perturbations of BMP/TGF-Î² and VEGF/VEGFR signalling pathways in non-syndromic sporadic brain arteriovenous malformations (BAVM). Journal of Medical Genetics, 2018, 55, 675-684.	1.5	70
81	Whole-exome sequencing reveals known and novel variants in a cohort of intracranial vertebralâ€“basilar artery dissection (IVAD). Journal of Human Genetics, 2018, 63, 1119-1128.	1.1	21
82	The next generation of population-based spinal muscular atrophy carrier screening: comprehensive pan-ethnic SMN1 copy-number and sequence variant analysis by massively parallel sequencing. Genetics in Medicine, 2017, 19, 936-944.	1.1	70
83	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	13.5	66
84	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
85	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	13.9	565
86	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
87	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	3.6	39
88	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	3.6	50
89	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	2.6	112
90	Nonrecurrent PMP22-RAI1 contiguous gene deletions arise from replication-based mechanisms and result in Smithâ€“Magenis syndrome with evident peripheral neuropathy. Human Genetics, 2016, 135, 1161-1174.	1.8	4

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91	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	2.6	45
92	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	5.8	227
93	De Novo Mutations in CHD4 , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. American Journal of Human Genetics, 2016, 99, 934-941.	2.6	111
94	Clinical characteristics and prognosis of 272 postterm choriocarcinoma patients at Peking Union Medical College Hospital: a retrospective cohort study. BMC Cancer, 2016, 16, 347.	1.1	15
95	Copy number analysis of the low-copy repeats at the primate NPHP1 locus by array comparative genomic hybridization. Genomics Data, 2016, 8, 106-109.	1.3	1
96	Aggressive tumor growth and clinical evolution in a patient with X-linked acro-gigantism syndrome. Endocrine, 2016, 51, 236-244.	1.1	45
97	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	2.6	98
98	Somatic mosaicism underlies X-linked acrogigantism syndrome in sporadic male subjects. Endocrine-Related Cancer, 2016, 23, 221-233.	1.6	75
99	Comparative Genomic Analyses of the Human NPHP1 Locus Reveal Complex Genomic Architecture and Its Regional Evolution in Primates. PLoS Genetics, 2015, 11, e1005686.	1.5	21
100	TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	13.9	239
101	X-linked acrogigantism syndrome: clinical profile and therapeutic responses. Endocrine-Related Cancer, 2015, 22, 353-367.	1.6	151
102	Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. American Journal of Human Genetics, 2015, 96, 555-564.	2.6	45
103	Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The PMP22-RAI1 Contiguous Gene Duplication Syndrome. American Journal of Human Genetics, 2015, 97, 691-707.	2.6	33
104	Gigantism and Acromegaly Due to Xq26 Microduplications and GPR101 Mutation. New England Journal of Medicine, 2014, 371, 2363-2374.	13.9	292
105	Inherited dup(17)(p11.2p11.2): Expanding the phenotype of the Potockiâ€“Lupski syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 500-504.	0.7	29
106	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	3.8	1,171
107	Mechanism, Prevalence, and More Severe Neuropathy Phenotype of the Charcot-Marie-Tooth Type 1A Triplication. American Journal of Human Genetics, 2014, 94, 462-469.	2.6	42
108	Passage Number is a Major Contributor to Genomic Structural Variations in Mouse iPSCs. Stem Cells, 2014, 32, 2657-2667.	1.4	40

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109	Lysinuric protein intolerance presenting with multiple fractures. Molecular Genetics and Metabolism Reports, 2014, 1, 176-183.	0.4	20
110	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	2.6	80
111	Replicative mechanisms of CNV formation preferentially occur as intrachromosomal events: evidence from Potockiâ€“Lupski duplication syndrome. Human Molecular Genetics, 2013, 22, 749-756.	1.4	13
112	Cardiovascular findings in duplication 17p11.2 syndrome. Genetics in Medicine, 2012, 14, 90-94.	1.1	32
113	Identification of the first recurrent PAR1 deletion in LÃ©ri-Weill dyschondrosteosis and idiopathic short stature reveals the presence of a novelSHOXenhancer. Journal of Medical Genetics, 2012, 49, 442-450.	1.5	63
114	Mechanisms for recurrent and complex human genomic rearrangements. Current Opinion in Genetics and Development, 2012, 22, 211-220.	1.5	289
115	Multiple de novo copy number variations in two subjects with developmental problems and multiple congenital anomalies. BMC Proceedings, 2012, 6, .	1.8	1
116	Inverted genomic segments and complex triplication rearrangements are mediated by inverted repeats in the human genome. Nature Genetics, 2011, 43, 1074-1081.	9.4	184
117	Chromosome Catastrophes Involve Replication Mechanisms Generating Complex Genomic Rearrangements. Cell, 2011, 146, 889-903.	13.5	391
118	Frequency of Nonallelic Homologous Recombination Is Correlated with Length of Homology: Evidence that Ectopic Synapsis Precedes Ectopic Crossing-Over. American Journal of Human Genetics, 2011, 89, 580-588.	2.6	104
119	Alu-specific microhomology-mediated deletion of the final exon of SPAST in three unrelated subjects with hereditary spastic paraplegia. Genetics in Medicine, 2011, 13, 582-592.	1.1	53
120	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Human Molecular Genetics, 2011, 20, 1975-1988.	1.4	74
121	Identification of Uncommon Recurrent Potocki-Lupski Syndrome-Associated Duplications and the Distribution of Rearrangement Types and Mechanisms in PTLS. American Journal of Human Genetics, 2010, 86, 462-470.	2.6	79
122	Mechanisms for Nonrecurrent Genomic Rearrangements Associated with CMT1A or HNPP: Rare CNVs as a Cause for Missing Heritability. American Journal of Human Genetics, 2010, 86, 892-903.	2.6	125
123	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341.	1.5	447
124	Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. Human Molecular Genetics, 2009, 18, 2188-2203.	1.4	165