

Pengfei Liu

List of Publications by Year in descending order

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Version: 2024-02-01

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	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	3.8	1,171
2	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	13.9	565
3	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
4	Chromosome Catastrophes Involve Replication Mechanisms Generating Complex Genomic Rearrangements. Cell, 2011, 146, 889-903.	13.5	391
5	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
6	Gigantism and Acromegaly Due to Xq26 Microduplications and <i>GPR101</i> Mutation. New England Journal of Medicine, 2014, 371, 2363-2374.	13.9	292
7	Mechanisms for recurrent and complex human genomic rearrangements. Current Opinion in Genetics and Development, 2012, 22, 211-220.	1.5	289
8	<i>TBX6</i> Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	13.9	239
9	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	5.8	227
10	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
11	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
12	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
13	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
14	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
15	X-linked acrogigantism syndrome: clinical profile and therapeutic responses. Endocrine-Related Cancer, 2015, 22, 353-367.	1.6	151
16	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
17	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	2.6	112
18	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

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19	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. <i>Genome Medicine</i> , 2018, 10, 74.	3.6	105
20	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. <i>Nature Communications</i> , 2020, 11, 4932.	5.8	105
21	Frequency of Nonallelic Homologous Recombination Is Correlated with Length of Homology: Evidence that Ectopic Synapsis Precedes Ectopic Crossing-Over. <i>American Journal of Human Genetics</i> , 2011, 89, 580-588.	2.6	104
22	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. <i>American Journal of Human Genetics</i> , 2016, 98, 347-357.	2.6	98
23	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	87
24	Loss of tubulin deglutamylase <i>CCP1</i> causes infantile-onset neurodegeneration. <i>EMBO Journal</i> , 2018, 37, .	3.5	86
25	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 745-754.	2.6	80
26	Identification of Uncommon Recurrent Potocki-Lupski Syndrome-Associated Duplications and the Distribution of Rearrangement Types and Mechanisms in PTLs. <i>American Journal of Human Genetics</i> , 2010, 86, 462-470.	2.6	79
27	Somatic mosaicism underlies X-linked acrogerontism syndrome in sporadic male subjects. <i>Endocrine-Related Cancer</i> , 2016, 23, 221-233.	1.6	75
28	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. <i>Human Molecular Genetics</i> , 2011, 20, 1975-1988.	1.4	74
29	Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. <i>Cell</i> , 2019, 176, 1310-1324.e10.	13.5	73
30	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. <i>Genetics in Medicine</i> , 2017, 19, 936-944.	1.1	70
31	Perturbations of BMP/TGF- β and VEGF/VEGFR signalling pathways in non-syndromic sporadic brain arteriovenous malformations (BAVM). <i>Journal of Medical Genetics</i> , 2018, 55, 675-684.	1.5	70
32	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. <i>Npj Genomic Medicine</i> , 2020, 5, 47.	1.7	67
33	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. <i>Cell</i> , 2017, 168, 830-842.e7.	13.5	66
34	Phenotypic expansion in <i>DDX3X</i> â€” a common cause of intellectual disability in females. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 1277-1285.	1.7	66
35	Identification of the first recurrent PAR1 deletion in LÃ©ry-Weill dyschondrosteosis and idiopathic short stature reveals the presence of a novel <i>SHOX</i> enhancer. <i>Journal of Medical Genetics</i> , 2012, 49, 442-450.	1.5	63
36	TBX6-associated congenital scoliosis (TACS) as a clinically distinguishable subtype of congenital scoliosis: further evidence supporting the compound inheritance and TBX6 gene dosage model. <i>Genetics in Medicine</i> , 2019, 21, 1548-1558.	1.1	60

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37	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. <i>Human Genetics</i> , 2018, 137, 553-567.	1.8	57
38	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2018, 103, 154-162.	2.6	56
39	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. <i>Genome Medicine</i> , 2019, 11, 48.	3.6	55
40	Loss of function of NCOR1 and NCOR2 impairs memory through a novel GABAergic hypothalamusâ€“CA3 projection. <i>Nature Neuroscience</i> , 2019, 22, 205-217.	7.1	54
41	Alu-specific microhomology-mediated deletion of the final exon of SPAST in three unrelated subjects with hereditary spastic paraplegia. <i>Genetics in Medicine</i> , 2011, 13, 582-592.	1.1	53
42	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. <i>Genetics in Medicine</i> , 2019, 21, 663-675.	1.1	52
43	Identification of novel candidate disease genes from de novo exonic copy number variants. <i>Genome Medicine</i> , 2017, 9, 83.	3.6	50
44	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 63.	3.6	50
45	Validation Studies for Single Circulating Trophoblast Genetic Testing as a Form of Noninvasive Prenatal Diagnosis. <i>American Journal of Human Genetics</i> , 2019, 105, 1262-1273.	2.6	47
46	Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. <i>American Journal of Human Genetics</i> , 2015, 96, 555-564.	2.6	45
47	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. <i>American Journal of Human Genetics</i> , 2016, 99, 720-727.	2.6	45
48	Aggressive tumor growth and clinical evolution in a patient with X-linked acro-gigantism syndrome. <i>Endocrine</i> , 2016, 51, 236-244.	1.1	45
49	Review of the phenotypic spectrum associated with haploinsufficiency of <i>MYRF1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1376-1382.	0.7	44
50	Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.	1.1	44
51	Mechanism, Prevalence, and More Severe Neuropathy Phenotype of the Charcot-Marie-Tooth Type 1A Triplication. <i>American Journal of Human Genetics</i> , 2014, 94, 462-469.	2.6	42
52	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. <i>Genome Medicine</i> , 2019, 11, 30.	3.6	42
53	Perturbations of genes essential for Müllerian duct and Wolffian duct development in Mayer-Rokitansky-Küster-Hauser syndrome. <i>American Journal of Human Genetics</i> , 2021, 108, 337-345.	2.6	41
54	Passage Number is a Major Contributor to Genomic Structural Variations in Mouse iPSCs. <i>Stem Cells</i> , 2014, 32, 2657-2667.	1.4	40

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55	Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). <i>Journal of Medical Genetics</i> , 2021, 58, 41-47.	1.5	40
56	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. <i>Genome Medicine</i> , 2017, 9, 73.	3.6	39
57	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. <i>American Journal of Human Genetics</i> , 2019, 105, 384-394.	2.6	37
58	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. <i>Genetics in Medicine</i> , 2020, 22, 1633-1641.	1.1	36
59	Disruptive variants of <i>CSDE1</i> associate with autism and interfere with neuronal development and synaptic transmission. <i>Science Advances</i> , 2019, 5, eaax2166.	4.7	35
60	Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The PMP22-RAI1 Contiguous Gene Duplication Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 691-707.	2.6	33
61	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. <i>Brain</i> , 2020, 143, 112-130.	3.7	33
62	Cardiovascular findings in duplication 17p11.2 syndrome. <i>Genetics in Medicine</i> , 2012, 14, 90-94.	1.1	32
63	A Genocentric Approach to Discovery of Mendelian Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 974-986.	2.6	30
64	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. <i>Genetics in Medicine</i> , 2020, 22, 1768-1776.	1.1	30
65	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 346-356.	2.6	30
66	Inherited dup(17)(p11.2p11.2): Expanding the phenotype of the Potocki-Lupski syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 500-504.	0.7	29
67	SGLT2 Inhibitors for Treatment of Refractory Hypomagnesemia: A Case Report of 3 Patients. <i>Kidney Medicine</i> , 2020, 2, 359-364.	1.0	29
68	<i>TBX6</i> missense variants expand the mutational spectrum in a non-Mendelian inheritance disease. <i>Human Mutation</i> , 2020, 41, 182-195.	1.1	27
69	Bi-allelic Mutations in NADSYN1 Cause Multiple Organ Defects and Expand the Genotypic Spectrum of Congenital NAD Deficiency Disorders. <i>American Journal of Human Genetics</i> , 2020, 106, 129-136.	2.6	27
70	Recurrent arginine substitutions in the <i>ACTG2</i> gene are the primary driver of disease burden and severity in visceral myopathy. <i>Human Mutation</i> , 2020, 41, 641-654.	1.1	27
71	Clinical exome sequencing data reveal high diagnostic yields for congenital diaphragmatic hernia plus (CDH+) and new phenotypic expansions involving CDH. <i>Journal of Medical Genetics</i> , 2022, 59, 270-278.	1.5	27
72	De novo missense variant in the GTPase effector domain (GED) of <i>DNM1L</i> leads to static encephalopathy and seizures. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003673.	0.5	24

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73	Predominant and novel de novo variants in 29 individuals with <i>ALG13</i> deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1333-1348.	1.7	24
74	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smith-Magenis syndrome. <i>Genome Medicine</i> , 2019, 11, 12.	3.6	23
75	De Novo Variants in CDK19 Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2020, 106, 717-725.	2.6	23
76	Increased TBX6 gene dosages induce congenital cervical vertebral malformations in humans and mice. <i>Journal of Medical Genetics</i> , 2020, 57, 371-379.	1.5	23
77	Comparative Genomic Analyses of the Human NPHP1 Locus Reveal Complex Genomic Architecture and Its Regional Evolution in Primates. <i>PLoS Genetics</i> , 2015, 11, e1005686.	1.5	21
78	Whole-exome sequencing reveals known and novel variants in a cohort of intracranial vertebral basilar artery dissection (IVAD). <i>Journal of Human Genetics</i> , 2018, 63, 1119-1128.	1.1	21
79	Exome sequencing reveals genetic architecture in patients with isolated or syndromic short stature. <i>Journal of Genetics and Genomics</i> , 2021, 48, 396-402.	1.7	21
80	Lysinuric protein intolerance presenting with multiple fractures. <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 176-183.	0.4	20
81	DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract. <i>Genetics in Medicine</i> , 2019, 21, 2755-2764.	1.1	19
82	Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in <i>FBN1</i> . <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1023.	0.6	19
83	Cell-based analysis of CAD variants identifies individuals likely to benefit from uridine therapy. <i>Genetics in Medicine</i> , 2020, 22, 1598-1605.	1.1	18
84	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. <i>Genetics in Medicine</i> , 2021, 23, 259-271.	1.1	18
85	Human and mouse studies establish TBX6 in Mendelian CAKUT and as a potential driver of kidney defects associated with the 16p11.2 microdeletion syndrome. <i>Kidney International</i> , 2020, 98, 1020-1030.	2.6	17
86	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. <i>Genetics in Medicine</i> , 2021, 23, 1075-1085.	1.1	16
87	Clinical characteristics and prognosis of 272 postterm choriocarcinoma patients at Peking Union Medical College Hospital: a retrospective cohort study. <i>BMC Cancer</i> , 2016, 16, 347.	1.1	15
88	Biallelic loss of function variants in <i>PPP1R21</i> cause a neurodevelopmental syndrome with impaired endocytic function. <i>Human Mutation</i> , 2019, 40, 267-280.	1.1	15
89	Biallelic and De Novo Variants in <i>DONSON</i> Reveal a Clinical Spectrum of Cell Cycleopathies with Microcephaly, Dwarfism and Skeletal Abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2056-2066.	0.7	15
90	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. <i>Expert Review of Molecular Diagnostics</i> , 2020, 20, 995-1002.	1.5	14

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91	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100074.	1.0	14
92	Replicative mechanisms of CNV formation preferentially occur as intrachromosomal events: evidence from Potocki-Lupski duplication syndrome. <i>Human Molecular Genetics</i> , 2013, 22, 749-756.	1.4	13
93	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate disease-associated loci for BAFopathies. <i>Genetics in Medicine</i> , 2022, 24, 364-373.	1.1	12
94	Expanding the FANCO/RAD51C associated phenotype: Cleft lip and palate and lobar holoprosencephaly, two rare findings in Fanconi anemia. <i>European Journal of Medical Genetics</i> , 2018, 61, 257-261.	0.7	11
95	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
96	Detection of low-level parental somatic mosaicism for clinically relevant SNVs and indels identified in a large exome sequencing dataset. <i>Human Genomics</i> , 2021, 15, 72.	1.4	11
97	Villoglandular adenocarcinoma of cervix: pathologic features, clinical management, and outcome. <i>Cancer Management and Research</i> , 2018, Volume 10, 3955-3961.	0.9	10
98	Germline mutation in <i>POLR2A</i> : a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100014.	1.0	10
99	Heterozygous variants in <i>SPTBN1</i> cause intellectual disability and autism. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2037-2045.	0.7	9
100	Phenotypic expansion in <i>KIF1A</i> -related dominant disorders: A description of novel variants and review of published cases. <i>Human Mutation</i> , 2020, 41, 2094-2104.	1.1	8
101	Identification and characterization of novel <i>ACD</i> variants: modulation of TPP1 protein level offsets the impact of germline loss-of-function variants on telomere length. <i>Journal of Physical Education and Sports Management</i> , 2021, 7, a005454.	0.5	8
102	Exome sequencing reveals a novel variant in <i>NFX1</i> causing intracranial aneurysm in a Chinese family. <i>Journal of NeuroInterventional Surgery</i> , 2020, 12, 221-226.	2.0	7
103	Analysis of the role of the human papillomavirus 16/18 E7 protein assay in screening for cervical intraepithelial neoplasia: a case control study. <i>BMC Cancer</i> , 2020, 20, 999.	1.1	7
104	Combined Genome Sequencing and RNA Analysis Reveals and Characterizes a Deep Intronic Variant in <i>IGHMBP2</i> in a Patient With Spinal Muscular Atrophy With Respiratory Distress Type 1. <i>Pediatric Neurology</i> , 2021, 114, 16-20.	1.0	7
105	Amino Acid-Level Signal-to-Noise Analysis Aids in Pathogenicity Prediction of Incidentally Identified <i>TTN</i> -Encoded Titin Truncating Variants. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003131.	1.6	7
106	Expanding the mutation and phenotype spectrum of <i>MYH3</i> -associated skeletal disorders. <i>Npj Genomic Medicine</i> , 2022, 7, 11.	1.7	7
107	De novo variants in <i>H3-3A</i> and <i>H3-3B</i> are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. <i>Npj Genomic Medicine</i> , 2021, 6, 104.	1.7	7
108	Clinical characterization of individuals with the distal 1q21.1 microdeletion. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1388-1398.	0.7	6

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109	Emerging technologies for prenatal diagnosis: The application of whole genome and RNA sequencing. <i>Prenatal Diagnosis</i> , 2022, 42, 686-696.	1.1	6
110	Biallelic variants in <i>WARS1</i> cause a highly variable neurodevelopmental syndrome and implicate a critical exon for normal auditory function. <i>Human Mutation</i> , 2022, 43, 1472-1489.	1.1	6
111	Genome sequencing analysis of a family with a child displaying severe abdominal distention and recurrent hypoglycemia. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1130.	0.6	5
112	Nonrecurrent PMP22-RAI1 contiguous gene deletions arise from replication-based mechanisms and result in Smith-Magenis syndrome with evident peripheral neuropathy. <i>Human Genetics</i> , 2016, 135, 1161-1174.	1.8	4
113	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 631428.	1.8	4
114	Expanding the phenotype, genotype and biochemical knowledge of <i>ALG3</i> . <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 987-1000.	1.7	4
115	Saturation mutagenesis defines novel mouse models of severe spine deformity. <i>DMM Disease Models and Mechanisms</i> , 2021, 14, .	1.2	4
116	Contribution of uniparental disomy in a clinical trio exome cohort of 2675 patients. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1792.	0.6	4
117	Genome sequencing reveals novel noncoding variants in <i>PLA2G6</i> and <i>LMNB1</i> causing progressive neurologic disease. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1892.	0.6	4
118	Monozygotic twins with non-Down syndrome associated <i>MLL</i> -rearranged hematologic malignancy and megakaryoblastic differentiation. <i>Leukemia and Lymphoma</i> , 2019, 60, 1083-1086.	0.6	3
119	Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples. <i>Journal of Genetic Counseling</i> , 2022, 31, 59-70.	0.9	3
120	A novel de novo intronic variant in <i>ITPR1</i> causes Gillespie syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2315-2324.	0.7	2
121	Multiple de novo copy number variations in two subjects with developmental problems and multiple congenital anomalies. <i>BMC Proceedings</i> , 2012, 6, .	1.8	1
122	Copy number analysis of the low-copy repeats at the primate <i>NPHP1</i> locus by array comparative genomic hybridization. <i>Genomics Data</i> , 2016, 8, 106-109.	1.3	1
123	Vertical transmission of a large calvarial ossification defect due to heterozygous variants of <i>ALX4</i> and <i>TWIST1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 916-922.	0.7	1
124	Front Cover, Volume 41, Issue 1. <i>Human Mutation</i> , 2020, 41, i.	1.1	0