List of Publications by Year in descending order

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337 papers	15,217 citations	³⁶³⁰³ 51 h-index	26613 107 g-index
343	343	343	23238
all docs	docs citations	times ranked	citing authors

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#	Article	IF	CITATIONS
1	Copy Number Variation in Human Health, Disease, and Evolution. Annual Review of Genomics and Human Genetics, 2009, 10, 451-481.	6.2	1,026
2	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
3	Whole-Genome Sequencing in a Patient with Charcot–Marie–Tooth Neuropathy. New England Journal of Medicine, 2010, 362, 1181-1191.	27.0	698
4	Cytosine, but not adenine, base editors induce genome-wide off-target mutations in rice. Science, 2019, 364, 292-295.	12.6	491
5	CRISPR/Cas9 for genome editing: progress, implications and challenges. Human Molecular Genetics, 2014, 23, R40-R46.	2.9	487
6	Non-coding genetic variants in human disease: Figure 1 Human Molecular Genetics, 2015, 24, R102-R110.	2.9	466
7	A single mutation in the prM protein of Zika virus contributes to fetal microcephaly. Science, 2017, 358, 933-936.	12.6	399
8	Genetic evidence supports demic diffusion of Han culture. Nature, 2004, 431, 302-305.	27.8	398
9	Chromosome Catastrophes Involve Replication Mechanisms Generating Complex Genomic Rearrangements. Cell, 2011, 146, 889-903.	28.9	391
10	The DNA replication FoSTeS/MMBIR mechanism can generate genomic, genic and exonic complex rearrangements in humans. Nature Genetics, 2009, 41, 849-853.	21.4	382
11	Human Pluripotent Stem Cell-Derived Neural Cells and Brain Organoids Reveal SARS-CoV-2 Neurotropism Predominates in Choroid Plexus Epithelium. Cell Stem Cell, 2020, 27, 937-950.e9.	11.1	314
12	Autism and other neuropsychiatric symptoms are prevalent in individuals with <i>MeCP2</i> duplication syndrome. Annals of Neurology, 2009, 66, 771-782.	5.3	271
13	Autism-like behaviours and germline transmission in transgenic monkeys overexpressing MeCP2. Nature, 2016, 530, 98-102.	27.8	260
14	Complex human chromosomal and genomic rearrangements. Trends in Genetics, 2009, 25, 298-307.	6.7	239
15	<i>TBX6</i> Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	27.0	239
16	Biallelic Mutations in CFAP43 and CFAP44 Cause Male Infertility with Multiple Morphological Abnormalities of the Sperm Flagella. American Journal of Human Genetics, 2017, 100, 854-864.	6.2	220
17	Increased LIS1 expression affects human and mouse brain development. Nature Genetics, 2009, 41, 168-177.	21.4	199
18	Ongoing global and regional adaptive evolution of SARS-CoV-2. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	196

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19	A chromosome-level genome of the spider <i>Trichonephila antipodiana</i> reveals the genetic basis of its polyphagy and evidence of an ancient whole-genome duplication event. GigaScience, 2021, 10, .	6.4	187
20	Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. Human Molecular Genetics, 2009, 18, 2188-2203.	2.9	165
21	Assessing structural variation in a personal genome—towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	2.8	153
22	Zinc-finger-nucleases mediate specific and efficient excision of HIV-1 proviral DNA from infected and latently infected human T cells. Nucleic Acids Research, 2013, 41, 7771-7782.	14.5	146
23	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. Nature Genetics, 2019, 51, 117-127.	21.4	144
24	Y chromosome evidence of earliest modern human settlement in East Asia and multiple origins of Tibetan and Japanese populations. BMC Biology, 2008, 6, 45.	3.8	129
25	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.	6.2	125
26	Bi-allelic Mutations in ARMC2 Lead to Severe Astheno-Teratozoospermia Due to Sperm Flagellum Malformations in Humans and Mice. American Journal of Human Genetics, 2019, 104, 331-340.	6.2	113
27	Bi-allelic DNAH8 Variants Lead to Multiple Morphological Abnormalities of the Sperm Flagella and Primary Male Infertility. American Journal of Human Genetics, 2020, 107, 330-341.	6.2	111
28	Loss-of-function mutations in QRICH2 cause male infertility with multiple morphological abnormalities of the sperm flagella. Nature Communications, 2019, 10, 433.	12.8	108
29	Genetic Structure of Hmong-Mien Speaking Populations in East Asia as Revealed by mtDNA Lineages. Molecular Biology and Evolution, 2005, 22, 725-734.	8.9	105
30	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.	6.2	104
31	The Nuclear Matrix Protein SAFB Cooperates with Major Satellite RNAs to Stabilize Heterochromatin Architecture Partially through Phase Separation. Molecular Cell, 2020, 77, 368-383.e7.	9.7	104
32	Rationally Designed APOBEC3B Cytosine Base Editors with Improved Specificity. Molecular Cell, 2020, 79, 728-740.e6.	9.7	104
33	Bi-allelic Mutations in TTC21A Induce Asthenoteratospermia in Humans and Mice. American Journal of Human Genetics, 2019, 104, 738-748.	6.2	103
34	The nuclear matrix protein HNRNPU maintains 3D genome architecture globally in mouse hepatocytes. Genome Research, 2018, 28, 192-202.	5.5	91
35	A <i>DNAH17</i> missense variant causes flagella destabilization and asthenozoospermia. Journal of Experimental Medicine, 2020, 217, .	8.5	88
36	X-Linked Congenital Hypertrichosis Syndrome Is Associated with Interchromosomal Insertions Mediated by a Human-Specific Palindrome near SOX3. American Journal of Human Genetics, 2011, 88, 819-826.	6.2	87

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37	The b2/b3 subdeletion shows higher risk of spermatogenic failure and higher frequency of complete AZFc deletion than the gr/gr subdeletion in a Chinese population. Human Molecular Genetics, 2009, 18, 1122-1130.	2.9	86
38	Partial deletions are associated with an increased risk of complete deletion in AZFc: a new insight into the role of partial AZFc deletions in male infertility. Journal of Medical Genetics, 2007, 44, 437-444.	3.2	82
39	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.	6.2	79
40	Highâ€resolution molecular genomic autopsy reveals complex sudden unexpected death in epilepsy risk profile. Epilepsia, 2014, 55, e6-12.	5.1	76
41	Deleterious variants in X-linked CFAP47 induce asthenoteratozoospermia and primary male infertility. American Journal of Human Genetics, 2021, 108, 309-323.	6.2	74
42	Microcephaly-Associated Protein WDR62 Regulates Neurogenesis through JNK1 in the Developing Neocortex. Cell Reports, 2014, 6, 104-116.	6.4	71
43	Bi-allelic Loss-of-function Variants in CFAP58 Cause Flagellar Axoneme and Mitochondrial Sheath Defects and Asthenoteratozoospermia in Humans and Mice. American Journal of Human Genetics, 2020, 107, 514-526.	6.2	71
44	Novel homozygous <i>CFAP69</i> mutations in humans and mice cause severe asthenoteratospermia with multiple morphological abnormalities of the sperm flagella. Journal of Medical Genetics, 2019, 56, 96-103.	3.2	70
45	Systematic revision of Entomobryidae (<scp>C</scp> ollembola) by integrating molecular and new morphological evidence. Zoologica Scripta, 2015, 44, 298-311.	1.7	67
46	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	28.9	66
47	Efficient typing of copy number variations in a segmental duplication-mediated rearrangement hotspot using multiplex competitive amplification. Journal of Human Genetics, 2012, 57, 545-551.	2.3	63
48	POSH Localizes Activated Rac1 to Control the Formation of Cytoplasmic Dilation of the Leading Process and Neuronal Migration. Cell Reports, 2012, 2, 640-651.	6.4	63
49	Identifying psychiatric disorder-associated gut microbiota using microbiota-related gene set enrichment analysis. Briefings in Bioinformatics, 2020, 21, 1016-1022.	6.5	63
50	Bi-allelic Mutations in TTC29 Cause Male Subfertility with Asthenoteratospermia in Humans and Mice. American Journal of Human Genetics, 2019, 105, 1168-1181.	6.2	62
51	Genetic studies of human diversity in East Asia. Philosophical Transactions of the Royal Society B: Biological Sciences, 2007, 362, 987-996.	4.0	61
52	Genomic disorders: A window into human gene and genome evolution. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 1765-1771.	7.1	60
53	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.	2.4	60
54	Phylogenomics from low overage wholeâ€genome sequencing. Methods in Ecology and Evolution, 2019, 10, 507-517.	5.2	59

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55	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.	3.8	57
56	Homozygous mutations in <i>SPEF2</i> induce multiple morphological abnormalities of the sperm flagella and male infertility. Journal of Medical Genetics, 2020, 57, 31-37.	3.2	57
57	Rare variants in FANCA induce premature ovarian insufficiency. Human Genetics, 2019, 138, 1227-1236.	3.8	56
58	Biallelic mutations of CFAP251 cause sperm flagellar defects and human male infertility. Journal of Human Genetics, 2019, 64, 49-54.	2.3	56
59	Biallelic mutations in <i>CFAP65</i> cause male infertility with multiple morphological abnormalities of the sperm flagella in humans and mice. Journal of Medical Genetics, 2020, 57, 89-95.	3.2	55
60	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.	2.4	53
61	Socioeconomic Deprivation Index Is Associated With Psychiatric Disorders: An Observational and Genome-wide Gene-by-Environment Interaction Analysis in the UK Biobank Cohort. Biological Psychiatry, 2021, 89, 888-895.	1.3	51
62	Disruption in <i>ACTL7A</i> causes acrosomal ultrastructural defects in human and mouse sperm as a novel male factor inducing early embryonic arrest. Science Advances, 2020, 6, eaaz4796.	10.3	50
63	Bi-allelic mutations of DNAH10 cause primary male infertility with asthenoteratozoospermia in humans and mice. American Journal of Human Genetics, 2021, 108, 1466-1477.	6.2	50
64	The Suppression of CRMP2 Expression by Bone Morphogenetic Protein (BMP)-SMAD Gradient Signaling Controls Multiple Stages of Neuronal Development. Journal of Biological Chemistry, 2010, 285, 39039-39050.	3.4	49
65	Molecular phylogeny reveals independent origins of body scales in Entomobryidae (Hexapoda:) Tj ETQq1 1 0.78	34314.rgBT 2.7	/Oyerlock 10
66	American Strain of Zika Virus Causes More Severe Microcephaly Than an Old Asian Strain in Neonatal Mice. EBioMedicine, 2017, 25, 95-105.	6.1	47
67	Epigenetic regulation of Atrophin1 by lysine-specific demethylase 1 is required for cortical progenitor maintenance. Nature Communications, 2014, 5, 5815.	12.8	46
68	TBX6 compound inheritance leads to congenital vertebral malformations in humans and mice. Human Molecular Genetics, 2019, 28, 539-547.	2.9	46
69	A spatial analysis of genetic structure of human populations in China reveals distinct difference between maternal and paternal lineages. European Journal of Human Genetics, 2008, 16, 705-717.	2.8	45
70	MacroH2A1 associates with nuclear lamina and maintains chromatin architecture in mouse liver cells. Scientific Reports, 2015, 5, 17186.	3.3	44
71	Genome mapping coupled with CRISPR gene editing reveals a P450 gene confers avermectin resistance in the beet armyworm. PLoS Genetics, 2021, 17, e1009680.	3.5	44
72	Identification of Copy Number Variation Hotspots in Human Populations. American Journal of Human Genetics, 2010, 87, 494-504.	6.2	42

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73	Molecular Analysis of a Deletion Hotspot in the NRXN1 Region Reveals the Involvement of Short Inverted Repeats in Deletion CNVs. American Journal of Human Genetics, 2013, 92, 375-386.	6.2	42
74	Genome-wide copy number variation study and gene expression analysis identify ABI3BP as a susceptibility gene for Kashin–Beck disease. Human Genetics, 2014, 133, 793-799.	3.8	42
75	Mechanism, Prevalence, and More Severe Neuropathy Phenotype of the Charcot-Marie-Tooth Type 1A Triplication. American Journal of Human Genetics, 2014, 94, 462-469.	6.2	42
76	Perturbations of genes essential for Müllerian duct and Wölffian duct development in Mayer-Rokitansky-Küster-Hauser syndrome. American Journal of Human Genetics, 2021, 108, 337-345.	6.2	41
77	Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). Journal of Medical Genetics, 2021, 58, 41-47.	3.2	40
78	<i>BRCA2</i> in Ovarian Development and Function. New England Journal of Medicine, 2019, 380, 1086-1087.	27.0	38
79	Potocki-Lupski Syndrome: A Microduplication Syndrome Associated with Oropharyngeal Dysphagia and Failure to Thrive. Journal of Pediatrics, 2011, 158, 655-659.e2.	1.8	36
80	Cryptic diversity, diversification and vicariance in two species complexes of <i><scp>T</scp>omocerus</i> (<scp>C</scp> ollembola, <scp>T</scp> omoceridae) from <scp>C</scp> hina. Zoologica Scripta, 2014, 43, 393-404.	1.7	36
81	A novel homozygous mutation in WDR19 induces disorganization of microtubules in sperm flagella and nonsyndromic asthenoteratospermia. Journal of Assisted Reproduction and Genetics, 2020, 37, 1431-1439.	2.5	35
82	Association of polymorphisms in four bilirubin metabolism genes with serum bilirubin in three Asian populations. Human Mutation, 2009, 30, 609-615.	2.5	34
83	Evaluation of copy number variation detection for a SNP array platform. BMC Bioinformatics, 2014, 15, 50.	2.6	34
84	Mea6 controls VLDL transport through the coordinated regulation of COPII assembly. Cell Research, 2016, 26, 787-804.	12.0	34
85	Genome-wide DNA methylation profile analysis identifies differentially methylated loci associated with ankylosis spondylitis. Arthritis Research and Therapy, 2017, 19, 177.	3.5	34
86	Additional genomic duplications in AZFc underlie the b2/b3 deletion-associated risk of spermatogenic impairment in Han Chinese population. Human Molecular Genetics, 2011, 20, 4411-4421.	2.9	33
87	NIPA2 located in 15q11.2 is mutated in patients with childhood absence epilepsy. Human Genetics, 2012, 131, 1217-1224.	3.8	33
88	A Novel c-Jun N-terminal Kinase (JNK) Signaling Complex Involved in Neuronal Migration during Brain Development. Journal of Biological Chemistry, 2016, 291, 11466-11475.	3.4	33
89	Species delimitation in the morphologically conserved <i>Coecobrya</i> (Collembola:) Tj ETQq1 1 0.784314 rgBT taxonomy. Zoologica Scripta, 2018, 47, 342-356.	/Overlock 1.7	10 Tf 50 1(33
90	Assessing the Relationship Between Gut Microbiota and Bone Mineral Density. Frontiers in Genetics, 2020, 11, 6.	2.3	33

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91	New species and rediagnosis of <i>Coecobrya</i> (Collembola: Entomobryidae), with a key to the species of the genus. Journal of Natural History, 2009, 43, 2597-2615.	0.5	32
92	Structural variation of the human genome: mechanisms, assays, and role in male infertility. Systems Biology in Reproductive Medicine, 2011, 57, 3-16.	2.1	32
93	<i>cTAGE5</i> deletion in pancreatic β cells impairs proinsulin trafficking and insulin biogenesis in mice. Journal of Cell Biology, 2017, 216, 4153-4164.	5.2	32
94	Deletion and duplication of 15q24: Molecular mechanisms and potential modification by additional copy number variants. Genetics in Medicine, 2010, 12, 573-586.	2.4	31
95	Homozygous loss-of-function mutations in FSIP2 cause male infertility with asthenoteratospermia. Journal of Genetics and Genomics, 2019, 46, 53-56.	3.9	31
96	A Genome-wide Expression Association Analysis Identifies Genes and Pathways Associated with Amyotrophic Lateral Sclerosis. Cellular and Molecular Neurobiology, 2018, 38, 635-639.	3.3	30
97	Screening for Differentially Expressed Circular RNAs in the Cartilage of Osteoarthritis Patients for Their Diagnostic Value. Genetic Testing and Molecular Biomarkers, 2019, 23, 706-716.	0.7	30
98	Patients with severe asthenoteratospermia carrying SPAC6 or RSPH3 mutations have a positive pregnancy outcome following intracytoplasmic sperm injection. Journal of Assisted Reproduction and Genetics, 2020, 37, 829-840.	2.5	30
99	A large-scale integrative analysis of GWAS and common meQTLs across whole life course identifies genes, pathways and tissue/cell types for three major psychiatric disorders. Neuroscience and Biobehavioral Reviews, 2018, 95, 347-352.	6.1	29
100	Screening of Duchenne Muscular Dystrophy (DMD) Mutations and Investigating Its Mutational Mechanism in Chinese Patients. PLoS ONE, 2014, 9, e108038.	2.5	28
101	Genome-Wide Gene Expression Analysis Suggests an Important Role of Hypoxia in the Pathogenesis of Endemic Osteochondropathy Kashin-Beck Disease. PLoS ONE, 2011, 6, e22983.	2.5	28
102	GJB1/Connexin 32 whole gene deletions in patients with X-linked Charcot–Marie–Tooth disease. Neurogenetics, 2010, 11, 465-470.	1.4	27
103	Molecular phylogeny supports S-chaetae as a key character better than jumping organs and body scales in classification of Entomobryoidea (Collembola). Scientific Reports, 2015, 5, 12471.	3.3	27
104	Comparative analysis of gene expression profiles of hip articular cartilage between non-traumatic necrosis and osteoarthritis. Gene, 2016, 591, 43-47.	2.2	27
105	A Genomewide Integrative Analysis of GWAS and eQTLs Data Identifies Multiple Genes and Gene Sets Associated with Obesity. BioMed Research International, 2018, 2018, 1-5.	1.9	27
106	Whole-exome sequencing reveals SALL4 variants in premature ovarian insufficiency: an update on genotype–phenotype correlations. Human Genetics, 2019, 138, 83-92.	3.8	27
107	<i>TBX6</i> missense variants expand the mutational spectrum in a nonâ€Mendelian inheritance disease. Human Mutation, 2020, 41, 182-195.	2.5	27
108	NovelCFAP43 andCFAP44 mutations cause male infertility with multiple morphological abnormalities of the sperm flagella (MMAF). Reproductive BioMedicine Online, 2019, 38, 769-778.	2.4	26

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109	Novel lossâ€ofâ€function variants in <scp><i>DNAH17</i></scp> cause multiple morphological abnormalities of the sperm flagella in humans and mice. Clinical Genetics, 2021, 99, 176-186.	2.0	26
110	Loss of DRC1 function leads to multiple morphological abnormalities of the sperm flagella and male infertility in human and mouse. Human Molecular Genetics, 2021, 30, 1996-2011.	2.9	26
111	New insight into the systematics of Tomoceridae (Hexapoda, Collembola) by integrating molecular and morphological evidence. Zoologica Scripta, 2016, 45, 286-299.	1.7	25
112	Integrating genome-wide association study and expression quantitative trait locus study identifies multiple genes and gene sets associated with schizophrenia. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2018, 81, 50-54.	4.8	24
113	RET somatic mutations are underrecognized in Hirschsprung disease. Genetics in Medicine, 2018, 20, 770-777.	2.4	24
114	Progress and perspective of <i>TBX6</i> gene in congenital vertebral malformations. Oncotarget, 2016, 7, 57430-57441.	1.8	24
115	Is It the Appropriate Time to Stop Applying Selenium Enriched Salt in Kashin-Beck Disease Areas in China?. Nutrients, 2015, 7, 6195-6212.	4.1	23
116	A TBX5 3′UTR variant increases the risk of congenital heart disease in the Han Chinese population. Cell Discovery, 2017, 3, 17026.	6.7	23
117	Bi-allelic truncating variants in CFAP206 cause male infertility in human and mouse. Human Genetics, 2021, 140, 1367-1377.	3.8	23
118	Increased genome instability in human DNA segments with self-chains: homology-induced structural variations via replicative mechanisms. Human Molecular Genetics, 2013, 22, 2642-2651.	2.9	22
119	Two novel copy number variations involving the α-globin gene cluster on chromosome 16 cause thalassemia in two Chinese families. Molecular Genetics and Genomics, 2016, 291, 1443-1450.	2.1	22
120	Imbalance of dietary nutrients and the associated differentially expressed genes and pathways may play important roles in juvenile Kashin-Beck disease. Journal of Trace Elements in Medicine and Biology, 2018, 50, 441-460.	3.0	22
121	Comparative analysis of gene expression profiles in normal hip human cartilage and cartilage from patients with necrosis of the femoral head. Arthritis Research and Therapy, 2016, 18, 98.	3.5	21
122	Pathogenesis and potential relative risk factors of diabetic neuropathic osteoarthropathy. Journal of Orthopaedic Surgery and Research, 2017, 12, 142.	2.3	21
123	Transcriptome-wide association study identifies susceptibility genes for rheumatoid arthritis. Arthritis Research and Therapy, 2021, 23, 38.	3.5	21
124	Genome-wide CRISPR screens reveal synthetic lethal interaction between CREBBP and EP300 in diffuse large B-cell lymphoma. Cell Death and Disease, 2021, 12, 419.	6.3	21
125	Genome-wide copy number variant analysis for congenital ventricular septal defects in Chinese Han population. BMC Medical Genomics, 2015, 9, 2.	1.5	20
126	Efficient CNV breakpoint analysis reveals unexpected structural complexity and correlation of dosage-sensitive genes with clinical severity in genomic disorders. Human Molecular Genetics, 2017, 26, 1927-1941.	2.9	20

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127	Bi-allelic SHOC1 loss-of-function mutations cause meiotic arrest and non-obstructive azoospermia. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-107042.	3.2	20
128	Homozygous mutations in <i>CCDC34</i> cause male infertility with oligoasthenoteratozoospermia in humans and mice. Journal of Medical Genetics, 2022, 59, 710-718.	3.2	20
129	Assessing the Associations of Blood Metabolites With Osteoporosis: A Mendelian Randomization Study. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 1850-1855.	3.6	19
130	Novel bi-allelic variants in DNAH2 cause severe asthenoteratozoospermia with multiple morphological abnormalities of the flagella. Reproductive BioMedicine Online, 2021, 42, 963-972.	2.4	19
131	cTACE5/MEA6 plays a critical role in neuronal cellular components trafficking and brain development. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E9449-E9458.	7.1	18
132	Integration of transcriptome-wide association study and messenger RNA expression profile to identify genes associated with osteoarthritis. Bone and Joint Research, 2020, 9, 130-138.	3.6	18
133	CFAP65 is required in the acrosome biogenesis and mitochondrial sheath assembly during spermiogenesis. Human Molecular Genetics, 2021, 30, 2240-2254.	2.9	18
134	Fifteen Novel EIF2B1-5 Mutations Identified in Chinese Children with Leukoencephalopathy with Vanishing White Matter and a Long Term Follow-Up. PLoS ONE, 2015, 10, e0118001.	2.5	17
135	Deletion of exon 4 in LAMA2 is the most frequent mutation in Chinese patients with laminin α2-related muscular dystrophy. Scientific Reports, 2018, 8, 14989.	3.3	17
136	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.	5.2	17
137	Genome-Wide Pathway Association Studies of Multiple Correlated Quantitative Phenotypes Using Principle Component Analyses. PLoS ONE, 2012, 7, e53320.	2.5	17
138	COL9A1 Gene Polymorphism Is Associated with Kashin-Beck Disease in a Northwest Chinese Han Population. PLoS ONE, 2015, 10, e0120365.	2.5	16
139	MAZ mediates the cross-talk between CT-1 and NOTCH1 signaling during gliogenesis. Scientific Reports, 2016, 6, 21534.	3.3	16
140	Clinical and molecular genetic analysis of a family with late-onset LAMA2-related muscular dystrophy. Brain and Development, 2016, 38, 242-249.	1.1	16
141	Comparison of microRNA expression profiles of Kashin-Beck disease, osteoarthritis and rheumatoid arthritis. Scientific Reports, 2017, 7, 540.	3.3	16
142	Delimiting species of Protaphorura (Collembola: Onychiuridae): integrative evidence based on morphology, DNA sequences and geography. Scientific Reports, 2017, 7, 8261.	3.3	16
143	An integrative analysis of transcriptome-wide association study and mRNA expression profile identified candidate genes for attention-deficit/hyperactivity disorder. Psychiatry Research, 2019, 282, 112639.	3.3	16
144	Assessing the effect of interaction between C-reactive protein and gut microbiome on the risks of anxiety and depression. Molecular Brain, 2021, 14, 133.	2.6	16

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145	CNV instability associated with DNA replication dynamics: evidence for replicative mechanisms in CNV mutagenesis. Human Molecular Genetics, 2015, 24, 1574-1583.	2.9	15
146	Genomeâ€wide association study and identification of chromosomal enhancer maps in multiple brain regions related to autism spectrum disorder. Autism Research, 2019, 12, 26-32.	3.8	15
147	Molecular phylogeny of Entomobrya (Collembola: Entomobryidae) from China: Color pattern groups and multiple origins. Insect Science, 2019, 26, 587-597.	3.0	15
148	Xq22 deletions and correlation with distinct neurological disease traits in females: Further evidence for a contiguous gene syndrome. Human Mutation, 2020, 41, 150-168.	2.5	15
149	Rare mutations in the autophagyâ€regulating gene <i>AMBRA1</i> contribute to human neural tube defects. Human Mutation, 2020, 41, 1383-1393.	2.5	15
150	Gut microbiota is associated with bone mineral density. Bone and Joint Research, 2021, 10, 734-741.	3.6	15
151	Exome-wide screening identifies novel rare risk variants for major depression disorder. Molecular Psychiatry, 2022, 27, 3069-3074.	7.9	15
152	Exome Sequencing and Functional Analysis Identifies a Novel Mutation in EXT1 Gene That Causes Multiple Osteochondromas. PLoS ONE, 2013, 8, e72316.	2.5	14
153	A peculiar cave species of Tomocerus (Collembola, Tomoceridae, Tomocerinae) from Vietnam, with a discussion of the postantennal organ andÂprelabralÂchaetae in Tomocerinae. ZooKeys, 2014, 408, 61-70.	1.1	14
154	Correlation between frequency of non-allelic homologous recombination and homology properties: evidence from homology-mediated CNV mutations in the human genome. Human Molecular Genetics, 2015, 24, 1225-1233.	2.9	14
155	Elevated microRNA-520d-5p in the serum of patients with Parkinson's disease, possibly through regulation of cereloplasmin expression. Neuroscience Letters, 2018, 687, 88-93.	2.1	14
156	Synergistic effect of selenium and UV-B radiation in enhancing antioxidant level of wheatgrass grown from selenium rich wheat. Journal of Food Biochemistry, 2018, 42, e12577.	2.9	14
157	Integrative analysis of genome-wide association study and brain region related enhancer maps identifies biological pathways for insomnia. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2018, 86, 180-185.	4.8	14
158	MircroRNA-10b Promotes Human Embryonic Stem Cell-Derived Cardiomyocyte Proliferation via Novel Target Gene LATS1. Molecular Therapy - Nucleic Acids, 2020, 19, 437-445.	5.1	14
159	New species of Homidia (Collembola, Entomobryidae) from eastern China with description of the first instar larvae. ZooKeys, 2011, 152, 21-42.	1.1	13
160	Replicative mechanisms of CNV formation preferentially occur as intrachromosomal events: evidence from Potocki–Lupski duplication syndrome. Human Molecular Genetics, 2013, 22, 749-756.	2.9	13
161	DAZ duplications confer the predisposition of Y chromosome haplogroup K* to non-obstructive azoospermia in Han Chinese populations. Human Reproduction, 2013, 28, 2440-2449.	0.9	13
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