

Nan Wu

List of Publications by Year in descending order

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Version: 2024-02-01

137
papers

4,860
citations

172457

29
h-index

123424

61
g-index

152
all docs

152
docs citations

152
times ranked

6773
citing authors

#	ARTICLE	IF	CITATIONS
1	ceRNA in cancer: possible functions and clinical implications. <i>Journal of Medical Genetics</i> , 2015, 52, 710-718.	3.2	1,031
2	<i>TBX6</i> Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. <i>New England Journal of Medicine</i> , 2015, 372, 341-350.	27.0	239
3	Identification of tumor immune infiltration-associated lncRNAs for improving prognosis and immunotherapy response of patients with non-small cell lung cancer. , 2020, 8, e000110.		239
4	Mechanical sensing protein PIEZO1 regulates bone homeostasis via osteoblast-osteoclast crosstalk. <i>Nature Communications</i> , 2020, 11, 282.	12.8	229
5	Reanalysis of Clinical Exome Sequencing Data. <i>New England Journal of Medicine</i> , 2019, 380, 2478-2480.	27.0	205
6	Insights into genetics, human biology and disease gleaned from family based genomic studies. <i>Genetics in Medicine</i> , 2019, 21, 798-812.	2.4	161
7	Piezo1/2 mediate mechanotransduction essential for bone formation through concerted activation of NFAT-YAP1- β -catenin. <i>ELife</i> , 2020, 9, .	6.0	161
8	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. <i>Nature Genetics</i> , 2019, 51, 117-127.	21.4	144
9	Recurrence-Associated Long Non-coding RNA Signature for Determining the Risk of Recurrence in Patients with Colon Cancer. <i>Molecular Therapy - Nucleic Acids</i> , 2018, 12, 518-529.	5.1	139
10	CRISPR/Cas9 in zebrafish: an efficient combination for human genetic diseases modeling. <i>Human Genetics</i> , 2017, 136, 1-12.	3.8	83
11	COVID-19 Quarantine Reveals That Behavioral Changes Have an Effect on Myopia Progression. <i>Ophthalmology</i> , 2021, 128, 1652-1654.	5.2	82
12	Association of angiotensin-converting enzyme 2 gene polymorphism and enzymatic activity with essential hypertension in different gender. <i>Medicine (United States)</i> , 2018, 97, e12917.	1.0	79
13	Perturbations of BMP/TGF- β 2 and VEGF/VEGFR signalling pathways in non-syndromic sporadic brain arteriovenous malformations (BAVM). <i>Journal of Medical Genetics</i> , 2018, 55, 675-684.	3.2	70
14	CD146 as a new marker for an increased chondroprogenitor cell subpopulation in the later stages of osteoarthritis. <i>Journal of Orthopaedic Research</i> , 2015, 33, 84-91.	2.3	69
15	Synovitis, acne, pustulosis, hyperostosis and osteitis syndrome: a single centre study of a cohort of 164 patients. <i>Rheumatology</i> , 2016, 55, 1023-1030.	1.9	67
16	FusionCancer: a database of cancer fusion genes derived from RNA-seq data. <i>Diagnostic Pathology</i> , 2015, 10, 131.	2.0	61
17	The Regulatory Roles of MicroRNAs in Bone Remodeling and Perspectives as Biomarkers in Osteoporosis. <i>BioMed Research International</i> , 2016, 2016, 1-11.	1.9	61
18	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.	2.4	60

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19	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.	3.8	57
20	Progress and Future Trends in PET/CT and PET/MRI Molecular Imaging Approaches for Breast Cancer. <i>Frontiers in Oncology</i> , 2020, 10, 1301.	2.8	55
21	The genetic landscape and clinical implications of vertebral anomalies in VACTERL association. <i>Journal of Medical Genetics</i> , 2016, 53, 431-437.	3.2	51
22	TBX6 compound inheritance leads to congenital vertebral malformations in humans and mice. <i>Human Molecular Genetics</i> , 2019, 28, 539-547.	2.9	46
23	Genetic Polymorphism of LBX1 Is Associated With Adolescent Idiopathic Scoliosis in Northern Chinese Han Population. <i>Spine</i> , 2017, 42, 1125-1129.	2.0	45
24	The Role of Semaphorin 3A in Bone Remodeling. <i>Frontiers in Cellular Neuroscience</i> , 2017, 11, 40.	3.7	44
25	Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.	2.4	44
26	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.	6.2	41
27	Spinal and sacroiliac involvement in SAPHO syndrome: A single center study of a cohort of 354 patients. <i>Seminars in Arthritis and Rheumatism</i> , 2019, 48, 990-996.	3.4	40
28	Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). <i>Journal of Medical Genetics</i> , 2021, 58, 41-47.	3.2	40
29	Vitamin D Receptor Activation in Liver Macrophages Protects Against Hepatic Endoplasmic Reticulum Stress in Mice. <i>Hepatology</i> , 2020, 71, 1453-1466.	7.3	38
30	ADAMTS5 in Osteoarthritis: Biological Functions, Regulatory Network, and Potential Targeting Therapies. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 703110.	3.5	34
31	A multi-ethnic meta-analysis confirms the association of rs6570507 with adolescent idiopathic scoliosis. <i>Scientific Reports</i> , 2018, 8, 11575.	3.3	33
32	CFEA: a cell-free epigenome atlas in human diseases. <i>Nucleic Acids Research</i> , 2020, 48, D40-D44.	14.5	32
33	Filamin B: The next hotspot in skeletal research?. <i>Journal of Genetics and Genomics</i> , 2017, 44, 335-342.	3.9	31
34	Genetic polymorphisms of <i>GPR126</i> are functionally associated with PUMC classifications of adolescent idiopathic scoliosis in a Northern Han population. <i>Journal of Cellular and Molecular Medicine</i> , 2018, 22, 1964-1971.	3.6	31
35	Genome-wide cell-free DNA methylation analyses improve accuracy of non-invasive diagnostic imaging for early-stage breast cancer. <i>Molecular Cancer</i> , 2021, 20, 36.	19.2	30
36	The involvement of ADAMTS5 genetic polymorphisms in predisposition and diffusion tensor imaging alterations of lumbar disc degeneration. <i>Journal of Orthopaedic Research</i> , 2014, 32, 686-694.	2.3	28

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37	Three patterns of osteoarticular involvement in SAPHO syndrome: a cluster analysis based on whole body bone scintigraphy of 157 patients. <i>Rheumatology</i> , 2019, 58, 1047-1055.	1.9	28
38	Integrative genomics analysis reveals a 21q22.11 locus contributing risk to COVID-19. <i>Human Molecular Genetics</i> , 2021, 30, 1247-1258.	2.9	28
39	<i>TBX6</i> missense variants expand the mutational spectrum in a non-Mendelian inheritance disease. <i>Human Mutation</i> , 2020, 41, 182-195.	2.5	27
40	Functional biology of the Steel syndrome founder allele and evidence for clan genomics derivation of COL27A1 pathogenic alleles worldwide. <i>European Journal of Human Genetics</i> , 2020, 28, 1243-1264.	2.8	27
41	Comparison of Apparent Diffusion Coefficient and T2 Relaxation Time Variation Patterns in Assessment of Age and Disc Level Related Intervertebral Disc Changes. <i>PLoS ONE</i> , 2013, 8, e69052.	2.5	26
42	Association between <i>ADAMTS-4</i> gene polymorphism and lumbar disc degeneration in Chinese Han population. <i>Journal of Orthopaedic Research</i> , 2016, 34, 860-864.	2.3	26
43	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. <i>Human Genetics</i> , 2018, 137, 689-703.	3.8	24
44	Progress and perspective of <i>TBX6</i> gene in congenital vertebral malformations. <i>Oncotarget</i> , 2016, 7, 57430-57441.	1.8	24
45	The Progress of CRISPR/Cas9-Mediated Gene Editing in Generating Mouse/Zebrafish Models of Human Skeletal Diseases. <i>Computational and Structural Biotechnology Journal</i> , 2019, 17, 954-962.	4.1	23
46	Increased <i>TBX6</i> gene dosages induce congenital cervical vertebral malformations in humans and mice. <i>Journal of Medical Genetics</i> , 2020, 57, 371-379.	3.2	23
47	Full-thickness excision using transanal endoscopic microsurgery for treatment of rectal neuroendocrine tumors. <i>World Journal of Gastroenterology</i> , 2015, 21, 9142.	3.3	23
48	Human antigen R enhances the epithelial-mesenchymal transition via regulation of ZEB-1 in the human airway epithelium. <i>Respiratory Research</i> , 2018, 19, 109.	3.6	22
49	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.	2.3	21
50	Exome sequencing reveals genetic architecture in patients with isolated or syndromic short stature. <i>Journal of Genetics and Genomics</i> , 2021, 48, 396-402.	3.9	21
51	An international meta-analysis confirms the association of BNC2 with adolescent idiopathic scoliosis. <i>Scientific Reports</i> , 2018, 8, 4730.	3.3	20
52	Association of ACE gene A2350G and I/D polymorphisms with essential hypertension in the northernmost province of China. <i>Clinical and Experimental Hypertension</i> , 2018, 40, 32-38.	1.3	20
53	Identification of novel FBN1 variations implicated in congenital scoliosis. <i>Journal of Human Genetics</i> , 2020, 65, 221-230.	2.3	20
54	Genetic polymorphisms of PAX1 are functionally associated with different PUMC types of adolescent idiopathic scoliosis in a northern Chinese Han population. <i>Gene</i> , 2019, 688, 215-220.	2.2	19

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55	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.	1.2	19
56	Whole-spine Computed Tomography Findings in SAPHO Syndrome. <i>Journal of Rheumatology</i> , 2017, 44, 648-654.	2.0	18
57	F-18 FDG PET/CT in 26 patients with SAPHO syndrome: a new vision of clinical and bone scintigraphy correlation. <i>Journal of Orthopaedic Surgery and Research</i> , 2018, 13, 120.	2.3	17
58	Whole-Genome Methylation Analysis of Phenotype Discordant Monozygotic Twins Reveals Novel Epigenetic Perturbation Contributing to the Pathogenesis of Adolescent Idiopathic Scoliosis. <i>Frontiers in Bioengineering and Biotechnology</i> , 2019, 7, 364.	4.1	17
59	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.	5.2	17
60	Comparative analysis of serum proteome in congenital scoliosis patients with <i>TBX6</i> haploinsufficiency – a first report pointing to lipid metabolism. <i>Journal of Cellular and Molecular Medicine</i> , 2018, 22, 533-545.	3.6	16
61	Clinical characteristics of 1,055 Chinese patients with Mayer-Rokitansky-Kuster-Hauser syndrome: a nationwide multicentric study. <i>Fertility and Sterility</i> , 2021, 116, 558-565.	1.0	16
62	Altered function in cartilage derived mesenchymal stem cell leads to OA-related cartilage erosion. <i>American Journal of Translational Research (discontinued)</i> , 2016, 8, 433-46.	0.0	16
63	Phenotypic and genetic spectrum of isolated macrodactyly: somatic mosaicism of PIK3CA and AKT1 oncogenic variants. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 288.	2.7	15
64	<i>KIAA1217</i> : A novel candidate gene associated with isolated and syndromic vertebral malformations. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1664-1672.	1.2	15
65	The mutational burden and oligogenic inheritance in Klippel-Feil syndrome. <i>BMC Musculoskeletal Disorders</i> , 2020, 21, 220.	1.9	15
66	IL-33 in the basolateral amygdala integrates neuroinflammation into anxiogenic circuits via modulating BDNF expression. <i>Brain, Behavior, and Immunity</i> , 2022, 102, 98-109.	4.1	15
67	Associations of SUCNR1, GRK4, CAMK1D gene polymorphisms and the susceptibility of type 2 diabetes mellitus and essential hypertension in a northern Chinese Han population. <i>Journal of Diabetes and Its Complications</i> , 2021, 35, 107752.	2.3	13
68	Possible Single-Nucleotide Polymorphism Loci Associated with Systemic Sclerosis Susceptibility: A Genetic Association Study in a Chinese Han Population. <i>PLoS ONE</i> , 2014, 9, e113197.	2.5	11
69	The genetic implication of scoliosis in osteogenesis imperfecta: a review. <i>Journal of Spine Surgery</i> , 2017, 3, 666-678.	1.2	11
70	Phenotypic heterogeneity of intellectual disability in patients with congenital insensitivity to pain with anhidrosis: A case report and literature review. <i>Journal of International Medical Research</i> , 2018, 46, 2445-2457.	1.0	11
71	A Recurrent Rare SOX9 Variant (M469V) is Associated with Congenital Vertebral Malformations. <i>Current Gene Therapy</i> , 2019, 19, 242-247.	2.0	11
72	Clinical characteristics of pediatric synovitis, acne, pustulosis, hyperostosis, and osteitis (SAPHO) syndrome: the first Chinese case series from a single center. <i>Clinical Rheumatology</i> , 2021, 40, 1487-1495.	2.2	11

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73	Small Heterodimer Partner (NR0B2) Coordinates Nutrient Signaling and the Circadian Clock in Mice. <i>Molecular Endocrinology</i> , 2016, 30, 988-995.	3.7	10
74	Progress and Application of CRISPR/Cas Technology in Biological and Biomedical Investigation. <i>Journal of Cellular Biochemistry</i> , 2017, 118, 3061-3071.	2.6	10
75	Efficacy of bisphosphonates in patients with synovitis, acne, pustulosis, hyperostosis, and osteitis syndrome: a prospective open study. <i>Clinical and Experimental Rheumatology</i> , 2019, 37, 663-669.	0.8	10
76	Recent Advances in Technique and Clinical Outcomes of Minimally Invasive Spine Surgery in Adult Scoliosis. <i>Chinese Medical Journal</i> , 2017, 130, 2608-2615.	2.3	9
77	Association of angiotensin-converting enzyme gene polymorphism with pulse pressure and its interaction with obesity status in Heilongjiang province. <i>Clinical and Experimental Hypertension</i> , 2019, 41, 70-74.	1.3	9
78	Demographic, clinical, and scintigraphic comparison of patients affected by palmoplantar pustulosis and severe acne: a retrospective study. <i>Clinical Rheumatology</i> , 2020, 39, 1989-1996.	2.2	8
79	Deciphering the mutational signature of congenital limb malformations. <i>Molecular Therapy - Nucleic Acids</i> , 2021, 24, 961-970.	5.1	8
80	Small airway remodeling in diabetic and smoking chronic obstructive pulmonary disease patients. <i>Aging</i> , 2020, 12, 7927-7944.	3.1	8
81	Association of LMX1A Genetic Polymorphisms With Susceptibility to Congenital Scoliosis in Chinese Han Population. <i>Spine</i> , 2014, 39, 1785-1791.	2.0	7
82	A novel haplotype of low-frequency variants in the aldosterone synthase gene among northern Han Chinese with essential hypertension. <i>Medicine (United States)</i> , 2017, 96, e8150.	1.0	7
83	Exome sequencing reveals a novel variant in NFX1 causing intracranial aneurysm in a Chinese family. <i>Journal of NeuroInterventional Surgery</i> , 2020, 12, 221-226.	3.3	7
84	Axial skeletal lesions and disease duration in SAPHO syndrome: A retrospective review of computed tomography findings in 81 patients. <i>International Journal of Rheumatic Diseases</i> , 2020, 23, 1152-1158.	1.9	7
85	Disruptive NADSYN1 Variants Implicated in Congenital Vertebral Malformations. <i>Genes</i> , 2021, 12, 1615.	2.4	7
86	Î22-microglobulin as a biomarker of pulmonary fibrosis development in COPD patients. <i>Aging</i> , 2021, 13, 1251-1263.	3.1	7
87	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. <i>Npj Genomic Medicine</i> , 2022, 7, 11.	3.8	7
88	De novo variants in H3-3A and H3-3B are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. <i>Npj Genomic Medicine</i> , 2021, 6, 104.	3.8	7
89	Novel <i>NTRK1</i> Frameshift Mutation in Congenital Insensitivity to Pain With Anhidrosis. <i>Journal of Child Neurology</i> , 2015, 30, 1357-1361.	1.4	6
90	Molecular therapeutic strategies for FGFR3 gene-related skeletal dysplasia. <i>Journal of Molecular Medicine</i> , 2017, 95, 1303-1313.	3.9	6

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91	Mutational burden and potential oligogenic model of <i>TBX6</i> -mediated genes in congenital scoliosis. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1453.	1.2	6
92	Whole-genome methylation analysis reveals novel epigenetic perturbations of congenital scoliosis. <i>Molecular Therapy - Nucleic Acids</i> , 2021, 23, 1281-1287.	5.1	6
93	Whole Exome Sequencing Uncovered the Genetic Architecture of Growth Hormone Deficiency Patients. <i>Frontiers in Endocrinology</i> , 2021, 12, 711991.	3.5	6
94	Insulin in high concentration recede cigarette smoke extract induced cellular senescence of airway epithelial cell through autophagy pathway. <i>Biochemical and Biophysical Research Communications</i> , 2019, 509, 498-505.	2.1	5
95	Comparative analysis of the two extremes of -mutated autosomal dominant disease spectrum: from clinical phenotypes to cellular and molecular findings. <i>American Journal of Translational Research (discontinued)</i> , 2018, 10, 1400-1412.	0.0	5
96	The Mutational Landscape of <i>PTK7</i> in Congenital Scoliosis and Adolescent Idiopathic Scoliosis. <i>Genes</i> , 2021, 12, 1791.	2.4	5
97	Construction of a circRNA-miRNA-mRNA Regulated Pathway Involved in EGFR-TKI Lung Adenocarcinoma Resistance. <i>Technology in Cancer Research and Treatment</i> , 2021, 20, 153303382110568.	1.9	5
98	Aberrant interaction between mutated <i>ADAMTSL2</i> and <i>LTBP4</i> is associated with adolescent idiopathic scoliosis. <i>Gene</i> , 2022, 814, 146126.	2.2	5
99	PhenoApt leverages clinical expertise to prioritize candidate genes via machine learning. <i>American Journal of Human Genetics</i> , 2022, 109, 270-281.	6.2	5
100	A genotype-first analysis in a cohort of Mullerian anomaly. <i>Journal of Human Genetics</i> , 2022, 67, 347-352.	2.3	5
101	A retrospective study of bone scintigraphy in the follow-up of patients with synovitis, acne, pustulosis, hyperostosis, and osteitis syndrome: is it useful to repeat bone scintigraphy for disease assessment?. <i>Clinical Rheumatology</i> , 2020, 39, 1305-1314.	2.2	4
102	Mutational landscape and genetic signatures of cell-free DNA in tumour-induced osteomalacia. <i>Journal of Cellular and Molecular Medicine</i> , 2020, 24, 4931-4943.	3.6	4
103	Heterozygous Recurrent Mutations Inducing Dysfunction of <i>ROR2</i> Gene in Patients With Short Stature. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 661747.	3.7	4
104	Advances in clinical genetics and genomics. <i>Intelligent Medicine</i> , 2021, 1, 128-133.	3.1	4
105	Adverse drug reactions of Yunnan Baiyao capsule: a multi-center intensive monitoring study in China. <i>Annals of Translational Medicine</i> , 2019, 7, 118-118.	1.7	4
106	Reinforced Laryngeal Mask in Pediatric Laparoscopic Surgery. <i>Journal of the College of Physicians and Surgeons-Pakistan: JCPSP</i> , 2019, 29, 915-918.	0.4	4
107	Serum IgG4 elevation in SAPHO syndrome: does it unmask a disease activity marker?. <i>Clinical and Experimental Rheumatology</i> , 2020, 38, 35-41.	0.8	4
108	DrABC: deep learning accurately predicts germline pathogenic mutation status in breast cancer patients based on phenotype data. <i>Genome Medicine</i> , 2022, 14, 21.	8.2	4

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109	Neurodevelopmental trajectory and modifiers of 16p11.2 microdeletion: A follow-up study of four Chinese children carriers. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1485.	1.2	3
110	Factors and predictive model associated with perioperative complications after long fusion in the treatment of adult non-degenerative scoliosis. <i>BMC Musculoskeletal Disorders</i> , 2021, 22, 483.	1.9	3
111	Response to Biesecker et al.. <i>American Journal of Human Genetics</i> , 2021, 108, 1807-1808.	6.2	3
112	Fat Mass and Obesity-Associated (FTO) Gene Polymorphisms Are Associated with Risk of Intervertebral Disc Degeneration in Chinese Han Population: A Case Control Study. <i>Medical Science Monitor</i> , 2018, 24, 5598-5609.	1.1	3
113	Evaluation of nestin or osterix promoter-driven cre/lox system in studying the biological functions of murine osteoblastic cells. <i>American Journal of Translational Research (discontinued)</i> , 2016, 8, 1447-59.	0.0	3
114	Activating PIK3CA postzygotic mutations in segmental overgrowth of muscles with bone involvement in the body extremities. <i>Molecular Genetics and Genomics</i> , 2022, 297, 387-396.	2.1	3
115	Prolonged Hoarseness Caused by Arytenoid Dislocation After Anterior Cervical Corpectomy and Fusion. <i>Spine</i> , 2016, 41, E174-E177.	2.0	2
116	Evaluation of a novel poly(amidoamine) with pendant aminobutyl group on the cellular properties of transfected bone marrow mesenchymal stem cells. <i>Journal of Biomedical Materials Research - Part A</i> , 2018, 106, 686-697.	4.0	2
117	Advances in understanding the genetics of syndromes involving congenital upper limb anomalies. <i>Annals of Joint</i> , 2019, 4, 30-30.	1.0	2
118	Cost-effectiveness analysis of using the TBX6-associated congenital scoliosis risk score (TACScore) in genetic diagnosis of congenital scoliosis. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 250.	2.7	2
119	A novel COMP mutation in a Chinese family with multiple epiphyseal dysplasia. <i>BMC Medical Genetics</i> , 2020, 21, 115.	2.1	2
120	Novel FGFR1 Variants Are Associated with Congenital Scoliosis. <i>Genes</i> , 2021, 12, 1126.	2.4	2
121	Estrogen Receptors (ESRs) Mutations in Adolescent Idiopathic Scoliosis: A Cross-Sectional Study. <i>Medical Science Monitor</i> , 2020, 26, e921611.	1.1	2
122	A single cohort, open-label study of the efficacy of pamidronate for palmoplantar pustulosis in synovitis, acne, pustulosis, hyperostosis and osteitis (SAPHO) syndrome. <i>Clinical and Experimental Rheumatology</i> , 2020, 38, 1263-1264.	0.8	2
123	Exome-wide Analysis of De Novo and Rare Genetic Variants in Patients With Brain Arteriovenous Malformation. <i>Neurology</i> , 2022, , 10.1212/WNL.0000000000200114.	1.1	2
124	Delineation of dual molecular diagnosis in patients with skeletal deformity. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 139.	2.7	2
125	Identification of variants in <i>ACAN</i> and <i>PAPSS2</i> leading to spondyloepi(meta)physeal dysplasias in four Chinese families. <i>Molecular Genetics & Genomic Medicine</i> , 2022, , e1916.	1.2	2
126	Diagnostic yield and clinical effects of exome sequencing analysis in patients with early-onset scoliosis. <i>Lancet, The</i> , 2019, 394, S79.	13.7	1

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127	SAT0359â€¦...THE AGREEMENT BETWEEN CT AND BONE SCINTIGRAPHY IN DETECTING OSTEOARTICULAR LESIONS IN SAPHO SYNDROME. , 2019, , .		1
128	A novel multiplex fluorescent competitive PCR for copy number variation detection. <i>Genomics</i> , 2019, 111, 1745-1751.	2.9	1
129	Mutational spectrum of syndromic genes in sporadic brain arteriovenous malformation. <i>Chinese Neurosurgical Journal</i> , 2022, 8, 4.	0.9	1
130	Identification of Novel FBN2 Variants in a Cohort of Congenital Contractural Arachnodactyly. <i>Frontiers in Genetics</i> , 2022, 13, 804202.	2.3	1
131	The Genetics Contributing to Disorders Involving Congenital Scoliosis. , 2018, , 89-106.		0
132	Front Cover, Volume 41, Issue 1. <i>Human Mutation</i> , 2020, 41, i.	2.5	0
133	Comparative proteomics analysis for identifying the lipid metabolism related pathways in patients with Klippel-Feil syndrome. <i>Annals of Translational Medicine</i> , 2021, 9, 255-255.	1.7	0
134	Variants Affecting the C-Terminal of CSF1R Cause Congenital Vertebral Malformation Through a Gain-of-Function Mechanism. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 641133.	3.7	0
135	Disease activity in patients with synovitis, acne, pustulosis, hyperostosis, and osteitis (SAPHO) syndrome: the utility of the SPARCC MRI scoring system for assessment of axial spine involvement. <i>Clinical and Experimental Rheumatology</i> , 2021, 39, 1291-1297.	0.8	0
136	The utility of hierarchical genetic testing in paediatric liver disease. <i>Liver International</i> , 2022, , .	3.9	0
137	The identification of PAX7 variants and a potential role of muscle development dysfunction in congenital scoliosis. <i>Cell Regeneration</i> , 2022, 11, 16.	2.6	0