Shiro Ikegawa

List of Publications by Year
in descending order

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323 papers	18,236 citations	14614 66 h-index	20307 116 g-index
340 all docs	340 docs citations	340 times ranked	21338 citing authors

#	Article	IF	Citations
1	Association Between Vitamin A Intake and Disease Severity in Early-Onset Heterotopic Ossification of the Posterior Longitudinal Ligament of the Spine. Global Spine Journal, 2022, 12, 1770-1780.	1.2	10
2	Genome-wide association study of colorectal polyps identified highly overlapping polygenic architecture with colorectal cancer. Journal of Human Genetics, 2022, 67, 149-156.	1.1	5
3	Novel susceptibility loci for steroid-associated osteonecrosis of the femoral head in systemic lupus erythematosus. Human Molecular Genetics, 2022, 31, 1082-1095.	1.4	1
4	Rare SLC13A1 variants associate with intervertebral disc disorder highlighting role of sulfate in disc pathology. Nature Communications, 2022, 13, 634.	5.8	21
5	SOX11 variants cause a neurodevelopmental disorder with infrequent ocular malformations and hypogonadotropic hypogonadism and with distinct DNA methylation profile. Genetics in Medicine, 2022, 24, 1261-1273.	1.1	14
6	De novo heterozygous variants in KIF5B cause kyphomelic dysplasia. Clinical Genetics, 2022, 102, 3-11.	1.0	5
7	The first study of epidemiology of adolescent idiopathic scoliosis shows lower prevalence in females of Jammu and Kashmir, India.. American Journal of Translational Research (discontinued), 2022, 14, 1100-1106.	0.0	1
8	Ossification of the posterior longitudinal ligament. , 2022, , 253-281.		0
9	The third case of TNFRSF11A-associated dysosteosclerosis with a mutation producing elongating proteins. Journal of Human Genetics, 2021, 66, 371-377.	1.1	8
10	Meta-analysis of 208370 East Asians identifies 113 susceptibility loci for systemic lupus erythematosus. Annals of the Rheumatic Diseases, 2021, 80, 632-640.	0.5	103
11	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	1.1	18
12	Genetic disorders associated with the RANKL/OPG/RANK pathway. Journal of Bone and Mineral Metabolism, 2021, 39, 45-53.	1.3	9
13	Differentiation of Hypertrophic Chondrocytes from Human iPSCs for the InÂVitro Modeling of Chondrodysplasias. Stem Cell Reports, 2021, 16, 610-625.	2.3	11
14	Deficiency of TMEM53 causes a previously unknown sclerosing bone disorder by dysregulation of BMP-SMAD signaling. Nature Communications, 2021, 12, 2046.	5.8	7
15	From HDLS to BANDDOS: fast-expanding phenotypic spectrum of disorders caused by mutations in CSF1R. Journal of Human Genetics, 2021, 66, 1139-1144.	1.1	15
16	Eight novel susceptibility loci and putative causal variants in atopic dermatitis. Journal of Allergy and Clinical Immunology, 2021, 148, 1293-1306.	1.5	32
17	Molecular Classification of Knee Osteoarthritis. Frontiers in Cell and Developmental Biology, 2021, 9, 725568.	1.8	38
18	CDC5L promotes early chondrocyte differentiation and proliferation by modulating pre-mRNA splicing of SOX9, COL2A1, and WEE1. Journal of Biological Chemistry, 2021, 297, 100994.	1.6	8

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19	Deciphering osteoarthritis genetics across 826,690 individuals from 9 populations. Cell, 2021, 184, 4784-4818.e17.	13.5	188
20	Expanding the phenotypic spectrum of TNFRSF11A-associated dysosteosclerosis: a case with intracranial extramedullary hematopoiesis. Journal of Human Genetics, 2021, 66, 607-611.	1.1	6
21	Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152.	1.7	26
22	Two unrelated pedigrees with achondrogenesis type 1b carrying a Japanâ€specific pathogenic variant in SLC26A2. American Journal of Medical Genetics, Part A, 2020, 182, 735-739.	0.7	0
23	Identification of novel FBN1 variations implicated in congenital scoliosis. Journal of Human Genetics, 2020, 65, 221-230.	1.1	20
24	A multi-ethnic meta-analysis identifies novel genes, including ACSL5, associated with amyotrophic lateral sclerosis. Communications Biology, 2020, 3, 526.	2.0	49
25	Large-scale genome-wide association study in a Japanese population identifies novel susceptibility loci across different diseases. Nature Genetics, 2020, 52, 669-679.	9.4	304
26	Recapitulating the human segmentation clock with pluripotent stem cells. Nature, 2020, 580, 124-129.	13.7	148
27	GWAS of 165,084 Japanese individuals identified nine loci associated with dietary habits. Nature Human Behaviour, 2020, 4, 308-316.	6.2	80
28	CANT1 deficiency in a mouse model of Desbuquois dysplasia impairs glycosaminoglycan synthesis and chondrocyte differentiation in growth plate cartilage. FEBS Open Bio, 2020, 10, 1096-1103.	1.0	10
29	Polygenic Risk Score of Adolescent Idiopathic Scoliosis for Potential Clinical Use. Journal of Bone and Mineral Research, 2020, 36, 1481-1491.	3.1	5
30	A Short History of the Genetic Study of OPLL. , 2020, , 55-60.		0
31	SLC4A2 Deficiency Causes a New Type of Osteopetrosis. Journal of Bone and Mineral Research, 2020, 37, 226-235.	3.1	12
32	Genome-wide association study identifies 14 previously unreported susceptibility loci for adolescent idiopathic scoliosis in Japanese. Nature Communications, 2019, 10, 3685.	5.8	47
33	Comprehensive clinical and molecular studies in split-hand/foot malformation: identification of two plausible candidate genes (LRP6 and UBA2). European Journal of Human Genetics, 2019, 27, 1845-1857.	1.4	11
34	Characterizing rare and low-frequency height-associated variants in the Japanese population. Nature Communications, 2019, 10, 4393.	5.8	123
35	TNFRSF11A-Associated Dysosteosclerosis: A Report of the Second Case and Characterization of the Phenotypic Spectrum. Journal of Bone and Mineral Research, 2019, 34, 1873-1879.	3.1	12
36	Bi-allelic loss of function variants ofTBX6causes a spectrum of malformation of spine and rib including congenital scoliosis and spondylocostal dysostosis. Journal of Medical Genetics, 2019, 56, 622-628.	1.5	13

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37	GWAS of smoking behaviour in 165,436 Japanese people reveals seven new loci and shared genetic architecture. Nature Human Behaviour, 2019, 3, 471-477.	6.2	54
38	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. American Journal of Human Genetics, 2019, 104, 925-935.	2.6	92
39	Meta-Analysis of Genome-Wide Association Studies Identifies Three Loci Associated With Stiffness Index of the Calcaneus. Journal of Bone and Mineral Research, 2019, 34, 1275-1283.	3.1	8
40	A multiethnic meta-analysis defined the association of rs12946942 with severe adolescent idiopathic scoliosis. Journal of Human Genetics, 2019, 64, 493-498.	1.1	11
41	Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. American Journal of Human Genetics, 2019, 104, 439-453.	2.6	16
42	Crim1C140S mutant mice reveal the importance of cysteine 140 in the internal region 1 of CRIM1 for its physiological functions. Mammalian Genome, 2019, 30, 329-338.	1.0	3
43	Association of Susceptibility Genes for Adolescent Idiopathic Scoliosis and Intervertebral Disc Degeneration With Adult Spinal Deformity. Spine, 2019, 44, 1623-1629.	1.0	13
44	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
45	Identification of novel LFNG mutations in spondylocostal dysostosis. Journal of Human Genetics, 2019, 64, 261-264.	1.1	17
46	A genomeâ€wide association study identifies new genes associated with developmental dysplasia of the hip. Clinical Genetics, 2019, 95, 345-355.	1.0	7
47	Identification of 28 new susceptibility loci for type 2 diabetes in the Japanese population. Nature Genetics, 2019, 51, 379-386.	9.4	164
48	Double non-contiguous fractures in a patient with spondylo-epiphyseal dysplasia with spinal ankylosis treated with open and percutaneous spinal fixation technique: a case report. BMC Research Notes, 2018, 11, 106.	0.6	1
49	Genetic analysis of quantitative traits in the Japanese population links cell types to complex human diseases. Nature Genetics, 2018, 50, 390-400.	9.4	613
50	Dysosteosclerosis is also caused by TNFRSF11A mutation. Journal of Human Genetics, 2018, 63, 769-774.	1.1	21
51	A screening method to distinguish syndromic from sporadic spinal extradural arachnoid cyst. Journal of Orthopaedic Science, 2018, 23, 455-458.	0.5	4
52	An international meta-analysis confirms the association of BNC2 with adolescent idiopathic scoliosis. Scientific Reports, 2018, 8, 4730.	1.6	20
53	A Replication Study for the Association of rs11190870 With Curve Severity in Adolescent Idiopathic Scoliosis in Japanese. Spine, 2018, 43, 688-692.	1.0	7
54	Genome-wide association study of knee osteoarthritis: present and future. Annals of Joint, 2018, 3, 64-64.	1.0	5

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55	Epigenetics for curve progression of adolescent idiopathic scoliosis. EBioMedicine, 2018, 37, 36-37.	2.7	9
56	Integrative genomic analysis for the functional roles of ITPKC in bone mineral density. Bioscience Reports, 2018, 38, .	1.1	1
57	Genome-wide meta-analysis and replication studies in multiple ethnicities identify novel adolescent idiopathic scoliosis susceptibility loci. Human Molecular Genetics, 2018, 27, 3986-3998.	1.4	34
58	Screening of known disease genes in congenital scoliosis. Molecular Genetics & Genomic Medicine, 2018, 6, 966-974.	0.6	20
59	A multi-ethnic meta-analysis confirms the association of rs6570507 with adolescent idiopathic scoliosis. Scientific Reports, 2018, 8, 11575.	1.6	33
60	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
61	Trans-Ethnic Polygenic Analysis Supports Genetic Overlaps of Lumbar Disc Degeneration With Height, Body Mass Index, and Bone Mineral Density. Frontiers in Genetics, 2018, 9, 267.	1.1	8
62	Further expansion of the mutational spectrum of spondylo-meta-epiphyseal dysplasia with abnormal calcification. Journal of Human Genetics, 2018, 63, 1003-1007.	1.1	8
63	Emergence of Zebrafish as a Model System for Understanding Human Scoliosis. , 2018, , 217-234.		2
64	Current Understanding of Genetic Factors in Idiopathic Scoliosis. , 2018, , 139-157.		0
65	Screening of the COL2A1 mutation in idiopathic osteonecrosis of the femoral head. Journal of Orthopaedic Research, 2017, 35, 768-774.	1.2	12
66	Axial spondylometaphyseal dysplasia is also caused by NEK1 mutations. Journal of Human Genetics, 2017, 62, 503-506.	1.1	25
67	Lumbar disc degeneration progression in young women in their 20's: A prospective ten-year follow up. Journal of Orthopaedic Science, 2017, 22, 635-640.	0.5	14
68	Identification of biallelic EXTL3 mutations in a novel type of spondylo-epi-metaphyseal dysplasia. Journal of Human Genetics, 2017, 62, 797-801.	1.1	35
69	Compound Heterozygosity for Null Mutations and a Common Hypomorphic Risk Haplotype inTBX6Causes Congenital Scoliosis. Human Mutation, 2017, 38, 317-323.	1.1	41
70	Novel and recurrent COL11A1 and COL2A1 mutations in the Marshallâ€“Stickler syndrome spectrum. Human Genome Variation, 2017, 4, 17040.	0.4	15
71	Genome-wide association study identifies 112 new loci for body mass index in the Japanese population. Nature Genetics, 2017, 49, 1458-1467.	9.4	380
72	Genome-wide Association Study of Idiopathic Osteonecrosis of the Femoral Head. Scientific Reports, 2017, 7, 15035.	1.6	23

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73	Identification of a novel LRRK1 mutation in a family with osteosclerotic metaphyseal dysplasia. Journal of Human Genetics, 2017, 62, 437-441.	1.1	33
74	Chondroitin SulfateN-acetylgalactosaminyltransferase-1 (CSGalNAcT-1) Deficiency Results in a Mild Skeletal Dysplasia and Joint Laxity. Human Mutation, 2017, 38, 34-38.	1.1	22
75	Novel and recurrent XYLT1 mutations in two Turkish families with Desbuquois dysplasia, type 2. Journal of Human Genetics, 2017, 62, 447-451.	1.1	24
76	An ENU-induced p.C225S missense mutation in the mouse Tgfb1 gene does not cause Camurati-Engelmann disease-like skeletal phenotypes. Experimental Animals, 2017, 66, 137-144.	0.7	2
77	A functional variant in MIR4300HG, the host gene of microRNA MIR4300 is associated with progression of adolescent idiopathic scoliosis. Human Molecular Genetics, 2017, 26, 4086-4092.	1.4	30
78	CELSR2 is a candidate susceptibility gene in idiopathic scoliosis. PLoS ONE, 2017, 12, e0189591.	1.1	17
79	Stickler Syndrome Type 1 with Short Stature and Atypical Ocular Manifestations. Case Reports in Pediatrics, 2016, 2016, 1-3.	0.2	7
80	Replication of Caucasian Loci Associated with Osteoporosis-related Traits in East Asians. Journal of Bone Metabolism, 2016, 23, 233.	0.5	9
81	Distinctive skeletal phenotype in high bone mass osteogenesis imperfecta due to a COL1A2 cleavage site mutation. American Journal of Medical Genetics, Part A, 2016, 170, 2212-2214.	0.7	5
82	Identification and Functional Characterization of RSPO2 as a Susceptibility Gene for Ossification of the Posterior Longitudinal Ligament of the Spine. American Journal of Human Genetics, 2016, 99, 202-207.	2.6	48
83	Identification of biallelicLRRK1mutations in osteosclerotic metaphyseal dysplasia and evidence for locus heterogeneity. Journal of Medical Genetics, 2016, 53, 568-574.	1.5	43
84	Deletions and de novo mutations ofSOX11are associated with a neurodevelopmental disorder with features of Coffinâ€“Siris syndrome. Journal of Medical Genetics, 2016, 53, 152-162.	1.5	69
85	Novel WISP3 mutations causing progressive pseudorheumatoid dysplasia in two Chinese families. Human Genome Variation, 2016, 3, 16041.	0.4	11
86	Whole Exome Screening Identifies Novel and Recurrent WISP3 Mutations Causing Progressive Pseudorheumatoid Dysplasia in Jammu and Kashmir-India. Scientific Reports, 2016, 6, 27684.	1.6	13
87	Identification of DNA methylation changes associated with disease progression in subchondral bone with site-matched cartilage in knee osteoarthritis. Scientific Reports, 2016, 6, 34460.	1.6	33
88	A novel type II collagen gene mutation in a family with spondyloepiphyseal dysplasia and extensive intrafamilial phenotypic diversity. Human Genome Variation, 2016, 3, 16007.	0.4	3
89	BGN Mutations in X-Linked Spondyloepimetaphyseal Dysplasia. American Journal of Human Genetics, 2016, 98, 1243-1248.	2.6	29
90	Genomic study of adolescent idiopathic scoliosis in Japan. Scoliosis and Spinal Disorders, 2016, 11, 5.	2.3	27

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91	Novel DDR2 mutation identified by whole exome sequencing in a Moroccan patient with spondyloâ€metaâ€epiphyseal dysplasia, short limbâ€abnormal calcification type. American Journal of Medical Genetics, Part A, 2016, 170, 460-465.	0.7	12
92	A rapid functional decline type of amyotrophic lateral sclerosis is linked to low expression ofTTN. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 851-858.	0.9	33
93	Functional Investigation of a Non-coding Variant Associated with Adolescent Idiopathic Scoliosis in Zebrafish: Elevated Expression of the Ladybird Homeobox Gene Causes Body Axis Deformation. PLoS Genetics, 2016, 12, e1005802.	1.5	51
94	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. PLoS ONE, 2016, 11, e0150555.	1.1	32
95	A novel FOXC2 mutation in spinal extradural arachnoid cyst. Human Genome Variation, 2015, 2, 15032.	0.4	11
96	Identification of HOXD4 Mutations in Spinal Extradural Arachnoid Cyst. PLoS ONE, 2015, 10, e0142126.	1.1	16
97	Influence of Intra-Articular Administration of Trichostatin A on Autologous Osteochondral Transplantation in a Rabbit Model. BioMed Research International, 2015, 2015, 1-8.	0.9	1
98	A PAX1 enhancer locus is associated with susceptibility to idiopathic scoliosis in females. Nature Communications, 2015, 6, 6452.	5.8	122
99	A Functional SNP in BNC2 Is Associated with Adolescent Idiopathic Scoliosis. American Journal of Human Genetics, 2015, 97, 337-342.	2.6	119
100	A novel CANT1 mutation in three Indian patients with Desbuquois dysplasia Kim type. European Journal of Medical Genetics, 2015, 58, 105-110.	0.7	12
101	A Case of Functional Growth Hormone Deficiency and Early Growth Retardation in a Child With IFT172 Mutations. Journal of Clinical Endocrinology and Metabolism, 2015, 100, 1221-1224.	1.8	29
102	Endoplasmic reticulum stress-mediated apoptosis contributes to a skeletal dysplasia resembling platyspondylic lethal skeletal dysplasia, Torrance type, in a novel Col2a1 mutant mouse line. Biochemical and Biophysical Research Communications, 2015, 468, 86-91.	1.0	12
103	Identification andIn VivoFunctional Characterization of Novel Compound HeterozygousBMP1Variants in Osteogenesis Imperfecta. Human Mutation, 2015, 36, 191-195.	1.1	25
104	Modeling type II collagenopathy skeletal dysplasia by directed conversion and induced pluripotent stem cells. Human Molecular Genetics, 2015, 24, 299-313.	1.4	35
105	Genetics of Ossification of the Posterior Longitudinal Ligament of the Spine: A Mini Review. Journal of Bone Metabolism, 2014, 21, 127.	0.5	29
106	A genome-wide association study identifies susceptibility loci for ossification of the posterior longitudinal ligament of the spine. Nature Genetics, 2014, 46, 1012-1016.	9.4	115
107	Severe manifestations of handâ€footâ€genital syndrome associated with a novel HOXA13 mutation. American Journal of Medical Genetics, Part A, 2014, 164, 2398-2402.	0.7	15
108	Cono-spondylar dysplasia: Clinical, radiographic, and molecular findings of a previously unreported disorder. , 2014, 164, 2147-2152.		0

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109	Molecular pathogenesis of Spondylocheirodysplastic Ehlersâ€Ðanlos syndrome caused by mutant ZIP13 proteins. EMBO Molecular Medicine, 2014, 6, 1028-1042.	3.3	56
110	Japanese founder duplications/triplications involving BHLHA9 are associated with split-hand/foot malformation with or without long bone deficiency and Gollop-Wolfgang complex. Orphanet Journal of Rare Diseases, 2014, 9, 125.	1.2	20
111	Mucopolysaccharidosis IVA (Morquio A syndrome) and VI (Maroteauxâ€“Lamy syndrome): under-recognized and challenging to diagnose. Skeletal Radiology, 2014, 43, 359-369.	1.2	39
112	De novo SOX11 mutations cause Coffinâ€“Siris syndrome. Nature Communications, 2014, 5, 4011.	5.8	118
113	Statin treatment rescues FGFR3 skeletal dysplasia phenotypes. Nature, 2014, 513, 507-511.	13.7	186
114	A meta-analysis identifies adolescent idiopathic scoliosis association withLBX1locus in multiple ethnic groups. Journal of Medical Genetics, 2014, 51, 401-406.	1.5	79
115	Cartilage intermediate layer protein promotes lumbar disc degeneration. Biochemical and Biophysical Research Communications, 2014, 446, 876-881.	1.0	27
116	Genomic study of ossification of the posterior longitudinal ligament of the spine. Proceedings of the Japan Academy Series B: Physical and Biological Sciences, 2014, 90, 405-412.	1.6	34
117	rs10865331 Associated with Susceptibility and Disease Severity of Ankylosing Spondylitis in a Taiwanese Population. PLoS ONE, 2014, 9, e104525.	1.1	11
118	The Genetics of Common Degenerative Skeletal Disorders: Osteoarthritis and Degenerative Disc Disease. Annual Review of Genomics and Human Genetics, 2013, 14, 245-256.	2.5	49
119	A genome-wide sib-pair linkage analysis of ossification of the posterior longitudinal ligament of the spine. Journal of Bone and Mineral Metabolism, 2013, 31, 136-143.	1.3	28
120	Mutations in B3GALT6, which Encodes a Glycosaminoglycan Linker Region Enzyme, Cause a Spectrum of Skeletal and Connective Tissue Disorders. American Journal of Human Genetics, 2013, 92, 927-934.	2.6	112
121	Genetic variants in GPR126 are associated with adolescent idiopathic scoliosis. Nature Genetics, 2013, 45, 676-679.	9.4	240
122	ZNF512B gene is a prognostic factor in patients with amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2013, 324, 163-166.	0.3	27
123	A Replication Study for Association of 5 Single Nucleotide Polymorphisms With Curve Progression of Adolescent Idiopathic Scoliosis in Japanese Patients. Spine, 2013, 38, 571-575.	1.0	23
124	A Replication Study for Association of 53 Single Nucleotide Polymorphisms in a Scoliosis Prognostic Test With Progression of Adolescent Idiopathic Scoliosis in Japanese. Spine, 2013, 38, 1375-1379.	1.0	28
125	Ectopic Expression of Ptf1a Induces Spinal Defects, Urogenital Defects, and Anorectal Malformations in Danforth's Short Tail Mice. PLoS Genetics, 2013, 9, e1003204.	1.5	17
126	Human Genetic Disorders Caused by Mutations in Genes Encoding Biosynthetic Enzymes for Sulfated Glycosaminoglycans*. Journal of Biological Chemistry, 2013, 288, 10953-10961.	1.6	93

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127	Meta-analysis identifies aMECOMgene as a novel predisposing factor of osteoporotic fracture. Journal of Medical Genetics, 2013, 50, 212-219.	1.5	30
128	Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and NEK2 as a new disease gene. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 16139-16144.	3.3	115
129	Osteogenesis imperfecta type V: Clinical and radiographic manifestations in mutation confirmed patients. American Journal of Medical Genetics, Part A, 2013, 161, 1972-1979.	0.7	27
130	Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused byPAPSS2Mutations. Human Mutation, 2013, 34, 1381-1386.	1.1	29
131	Exome sequencing identifies a novel INPPL1 mutation in opsismodysplasia. Journal of Human Genetics, 2013, 58, 391-394.	1.1	16
132	Association of the formiminotransferase N-terminal sub-domain containing gene and thrombospondin, type 1, domain-containing 7A gene with the prevalence of vertebral fracture in 2427 consecutive autopsy cases. Journal of Human Genetics, 2013, 58, 109-112.	1.1	3
133	Identification of a Susceptibility Locus for Severe Adolescent Idiopathic Scoliosis on Chromosome 17q24.3. PLoS ONE, 2013, 8, e72802.	1.1	59
134	FOXC2 Mutations in Familial and Sporadic Spinal Extradural Arachnoid Cyst. PLoS ONE, 2013, 8, e80548.	1.1	21
135	Lumbar disc degeneration is linked to a carbohydrate sulfotransferase 3 variant. Journal of Clinical Investigation, 2013, 123, 4909-4917.	3.9	126
136	Association Study of Polymorphisms rs4552569 and rs17095830 and the Risk of Ankylosing Spondylitis in a Taiwanese Population. PLoS ONE, 2013, 8, e52801.	1.1	10
137	TGF-Î² and Genetic Skeletal Diseases. , 2013, , 371-390.		0
138	Optineurin mutations in Japanese amyotrophic lateral sclerosis: Table 1. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 233-235.	0.9	35
139	SIK3 is essential for chondrocyte hypertrophy during skeletal development in mice. Development (Cambridge), 2012, 139, 1153-1163.	1.2	77
140	SNP rs11190870 near LBX1 is associated with adolescent idiopathic scoliosis in southern Chinese. Journal of Human Genetics, 2012, 57, 244-246.	1.1	64
141	PAPSS2mutations cause autosomal recessive brachyolmia. Journal of Medical Genetics, 2012, 49, 533-538.	1.5	44
142	Large-scale screening of TARDBP mutation in amyotrophic lateral sclerosis in Japanese. Neurobiology of Aging, 2012, 33, 786-790.	1.5	39
143	Novel deletion mutations of OPTN in amyotrophic lateral sclerosis in Japanese. Neurobiology of Aging, 2012, 33, 1843.e19-1843.e24.	1.5	37
144	Recurrence of osteogenesis imperfecta due to maternal mosaicism of a novel COL1A1 mutation. American Journal of Medical Genetics, Part A, 2012, 158A, 2969-2971.	0.7	2

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145	A Single Recurrent Mutation in the 5â€²-UTR of IFITM5 Causes Osteogenesis Imperfecta Type V. American Journal of Human Genetics, 2012, 91, 343-348.	2.6	216
146	A largeâ€scale replication study for the association of rs17039192 in HIFâ€2Î± with knee osteoarthritis. Journal of Orthopaedic Research, 2012, 30, 1244-1248.	1.2	14
147	TRPV4â€pathy manifesting both skeletal dysplasia and peripheral neuropathy: A report of three patients. American Journal of Medical Genetics, Part A, 2012, 158A, 795-802.	0.7	45
148	TRPV4â€associated skeletal dysplasias. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 190-204.	0.7	71
149	Genetic study on developmental dysplasia of the hip. European Journal of Clinical Investigation, 2012, 42, 1121-1125.	1.7	35
150	Disease-associated mutations in the actin-binding domain of filamin B cause cytoplasmic focal accumulations correlating with disease severity. Human Mutation, 2012, 33, 665-673.	1.1	31
151	Identification of a quantitative trait locus for spontaneous osteoarthritis in STR/ort mice. Journal of Orthopaedic Research, 2012, 30, 15-20.	1.2	6
152	A Short History of the Genome-Wide Association Study: Where We Were and Where We Are Going. Genomics and Informatics, 2012, 10, 220.	0.4	41
153	Association of the D repeat polymorphism in the ASPNgene with developmental dysplasia of the hip: a case-control study in Han Chinese. Arthritis Research and Therapy, 2011, 13, R27.	1.6	60
154	SNPs on chromosome 5p15.3 associated with myocardial infarction in Japanese population. Journal of Human Genetics, 2011, 56, 47-51.	1.1	41
155	Replication analysis of SNPs on 9p21.2 and 19p13.3 with amyotrophic lateral sclerosis in East Asians. Neurobiology of Aging, 2011, 32, 757.e13-757.e14.	1.5	23
156	Meta-analysis of genome-wide association studies confirms a susceptibility locus for knee osteoarthritis on chromosome 7q22. Annals of the Rheumatic Diseases, 2011, 70, 349-355.	0.5	126
157	Common Variants in a Novel Gene, FONG on Chromosome 2q33.1 Confer Risk of Osteoporosis in Japanese. PLoS ONE, 2011, 6, e19641.	1.1	35
158	Replication studies in various ethnic populations do not support the association of the HIF-2Î± SNP rs17039192 with knee osteoarthritis. Nature Medicine, 2011, 17, 26-27.	15.2	21
159	A genome-wide association study identifies common variants near LBX1 associated with adolescent idiopathic scoliosis. Nature Genetics, 2011, 43, 1237-1240.	9.4	233
160	SMOC1 Is Essential for Ocular and Limb Development in Humans and Mice. American Journal of Human Genetics, 2011, 88, 30-41.	2.6	100
161	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. American Journal of Human Genetics, 2011, 89, 767-772.	2.6	31
162	Cloning and characterization of the osteoarthritis-associated gene DVWA. Journal of Bone and Mineral Metabolism, 2011, 29, 300-308.	1.3	16

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163	ENU-induced missense mutation in the C-propeptide coding region of Col2a1 creates a mouse model of platyspondylic lethal skeletal dysplasia, Torrance type. Mammalian Genome, 2011, 22, 318-328.	1.0	18
164	Replication study of the association between adolescent idiopathic scoliosis and two estrogen receptor genes. Journal of Orthopaedic Research, 2011, 29, 834-837.	1.2	46
165	Lack of association between adolescent idiopathic scoliosis and previously reported single nucleotide polymorphisms in MATN1, MTNR1B, TPH1, and IGF1 in a Japanese population. Journal of Orthopaedic Research, 2011, 29, 1055-1058.	1.2	56
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