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	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
2	Identification of a novel non-coding RNA, MIAT, that confers risk of myocardial infarction. Journal of Human Genetics, 2006, 51, 1087-1099.	1.1	597
3	A functional polymorphism in the 5â€² UTR of GDF5 is associated with susceptibility to osteoarthritis. Nature Genetics, 2007, 39, 529-533.	9.4	435
4	An aspartic acid repeat polymorphism in asporin inhibits chondrogenesis and increases susceptibility to osteoarthritis. Nature Genetics, 2005, 37, 138-144.	9.4	424
5	Mutation in Npps in a mouse model of ossification of the posterior longitudinal ligament of the spine. Nature Genetics, 1998, 19, 271-273.	9.4	392
6	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
7	The combination of SOX5, SOX6, and SOX9 (the SOX trio) provides signals sufficient for induction of permanent cartilage. Arthritis and Rheumatism, 2004, 50, 3561-3573.	6.7	322
8	Gain-of-function mutations in TRPV4 cause autosomal dominant brachyolmia. Nature Genetics, 2008, 40, 999-1003.	9.4	320
9	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
10	Signalling mediated by the endoplasmic reticulum stress transducer OASIS is involved in bone formation. Nature Cell Biology, 2009, 11, 1205-1211.	4.6	278
11	A regulatory variant in CCR6 is associated with rheumatoid arthritis susceptibility. Nature Genetics, 2010, 42, 515-519.	9.4	241
12	Genetic variants in GPR126 are associated with adolescent idiopathic scoliosis. Nature Genetics, 2013, 45, 676-679.	9.4	240
13	The Zinc Transporter SLC39A13/ZIP13 Is Required for Connective Tissue Development; Its Involvement in BMP/TGF-Î² Signaling Pathways. PLoS ONE, 2008, 3, e3642.	1.1	240
14	Domain-specific mutations in TGFB1 result in Camurati-Engelmann disease. Nature Genetics, 2000, 26, 19-20.	9.4	239
15	A genome-wide association study identifies common variants near LBX1 associated with adolescent idiopathic scoliosis. Nature Genetics, 2011, 43, 1237-1240.	9.4	233
16	A functional SNP in CILP, encoding cartilage intermediate layer protein, is associated with susceptibility to lumbar disc disease. Nature Genetics, 2005, 37, 607-612.	9.4	223
17	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
18	Follistatin-like 1 (Fstl1) is a bone morphogenetic protein (BMP) 4 signaling antagonist in controlling mouse lung development. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 7058-7063.	3.3	197

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19	Identification of DIO2 as a new susceptibility locus for symptomatic osteoarthritis. Human Molecular Genetics, 2008, 17, 1867-1875.	1.4	190
20	Deciphering osteoarthritis genetics across 826,690 individuals from 9 populations. Cell, 2021, 184, 4784-4818.e17.	13.5	188
21	Statin treatment rescues FGFR3 skeletal dysplasia phenotypes. Nature, 2014, 513, 507-511.	13.7	186
22	Largeâ€scale analysis of association between GDF5 and FRZB variants and osteoarthritis of the hip, knee, and hand. Arthritis and Rheumatism, 2009, 60, 1710-1721.	6.7	181
23	Regulation of endoplasmic reticulum stress response by a BBF2H7-mediated Sec23a pathway is essential for chondrogenesis. Nature Cell Biology, 2009, 11, 1197-1204.	4.6	181
24	PLAP-1/Asporin, a Novel Negative Regulator of Periodontal Ligament Mineralization*. Journal of Biological Chemistry, 2007, 282, 23070-23080.	1.6	180
25	Identification of 28 new susceptibility loci for type 2 diabetes in the Japanese population. Nature Genetics, 2019, 51, 379-386.	9.4	164
26	Association of the human NPPS gene with ossification of the posterior longitudinal ligament of the spine (OPLL). Human Genetics, 1999, 104, 492-497.	1.8	159
27	A meta-analysis of European and Asian cohorts reveals a global role of a functional SNP in the 5' UTR of GDF5 with osteoarthritis susceptibility. Human Molecular Genetics, 2008, 17, 1497-1504.	1.4	156
28	Mechanisms for Asporin Function and Regulation in Articular Cartilage. Journal of Biological Chemistry, 2007, 282, 32185-32192.	1.6	151
29	Recapitulating the human segmentation clock with pluripotent stem cells. Nature, 2020, 580, 124-129.	13.7	148
30	The phenotypic spectrum ofCOL2A1mutations. Human Mutation, 2005, 26, 36-43.	1.1	146
31	A Functional Polymorphism in COL11A1, Which Encodes the Î±1 Chain of Type XI Collagen, Is Associated with Susceptibility to Lumbar Disc Herniation. American Journal of Human Genetics, 2007, 81, 1271-1277.	2.6	144
32	Genomewide Linkage and Linkage Disequilibrium Analyses Identify COL6A1, on Chromosome 21, as the Locus for Ossification of the Posterior Longitudinal Ligament of the Spine. American Journal of Human Genetics, 2003, 73, 812-822.	2.6	137
33	Loss-of-function mutations of CHST14 in a new type of Ehlers-Danlos syndrome. Human Mutation, 2010, 31, 966-974.	1.1	137
34	Common variants in DVWA on chromosome 3p24.3 are associated with susceptibility to knee osteoarthritis. Nature Genetics, 2008, 40, 994-998.	9.4	134
35	Association of the Asporin D14 Allele with Lumbar-Disc Degeneration in Asians. American Journal of Human Genetics, 2008, 82, 744-747.	2.6	132
36	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

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37	Lumbar disc degeneration is linked to a carbohydrate sulfotransferase 3 variant. Journal of Clinical Investigation, 2013, 123, 4909-4917.	3.9	126
38	Characterizing rare and low-frequency height-associated variants in the Japanese population. Nature Communications, 2019, 10, 4393.	5.8	123
39	Lethal Skeletal Dysplasia in Mice and Humans Lacking the Golgin GMAP-210. New England Journal of Medicine, 2010, 362, 206-216.	13.9	122
40	A PAX1 enhancer locus is associated with susceptibility to idiopathic scoliosis in females. Nature Communications, 2015, 6, 6452.	5.8	122
41	A Functional SNP in BNC2 Is Associated with Adolescent Idiopathic Scoliosis. American Journal of Human Genetics, 2015, 97, 337-342.	2.6	119
42	De novo SOX11 mutations cause Coffinâ€“Siris syndrome. Nature Communications, 2014, 5, 4011.	5.8	118
43	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
44	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
45	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
46	Distinct roles of Sox5, Sox6, and Sox9 in different stages of chondrogenic differentiation. Journal of Bone and Mineral Metabolism, 2005, 23, 337-340.	1.3	111
47	A functional single nucleotide polymorphism in the core promoter region of CALM1 is associated with hip osteoarthritis in Japanese. Human Molecular Genetics, 2005, 14, 1009-1017.	1.4	106
48	Loss-of-Function Mutations in PTPN11 Cause Metachondromatosis, but Not Ollier Disease or Maffucci Syndrome. PLoS Genetics, 2011, 7, e1002050.	1.5	104
49	Nucleotide-sugar transporter SLC35D1 is critical to chondroitin sulfate synthesis in cartilage and skeletal development in mouse and human. Nature Medicine, 2007, 13, 1363-1367.	15.2	103
50	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
51	A functional SNP in PSMA6 confers risk of myocardial infarction in the Japanese population. Nature Genetics, 2006, 38, 921-925.	9.4	102
52	A Functional Polymorphism in THBS2 that Affects Alternative Splicing and MMP Binding Is Associated with Lumbar-Disc Herniation. American Journal of Human Genetics, 2008, 82, 1122-1129.	2.6	102
53	SNPs in BRAP associated with risk of myocardial infarction in Asian populations. Nature Genetics, 2009, 41, 329-333.	9.4	102
54	SMOC1 Is Essential for Ocular and Limb Development in Humans and Mice. American Journal of Human Genetics, 2011, 88, 30-41.	2.6	100

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55	New Sequence Variants in HLA Class II/III Region Associated with Susceptibility to Knee Osteoarthritis Identified by Genome-Wide Association Study. PLoS ONE, 2010, 5, e9723.	1.1	96
56	A recurrent mutation in type II collagen gene causes Legg-CalvÃ©-Perthes disease in a Japanese family. Human Genetics, 2007, 121, 625-629.	1.8	95
57	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
58	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
59	Association of a single nucleotide polymorphism in growth differentiate factor 5 with congenital dysplasia of the hip: a case-control study. Arthritis Research and Therapy, 2008, 10, R126.	1.6	88
60	SOX9-dependent and -independent Transcriptional Regulation of Human Cartilage Link Protein. Journal of Biological Chemistry, 2004, 279, 50942-50948.	1.6	84
61	Replication of the association of the aspartic acid repeat polymorphism in the asporin gene with knee-osteoarthritis susceptibility in Han Chinese. Journal of Human Genetics, 2006, 51, 1068-1072.	1.1	80
62	GWAS of 165,084 Japanese individuals identified nine loci associated with dietary habits. Nature Human Behaviour, 2020, 4, 308-316.	6.2	80
63	A meta-analysis identifies adolescent idiopathic scoliosis association withLBX1locus in multiple ethnic groups. Journal of Medical Genetics, 2014, 51, 401-406.	1.5	79
64	Meta-analysis of association between the ASPN D-repeat and osteoarthritis. Human Molecular Genetics, 2007, 16, 1676-1681.	1.4	78
65	SIK3 is essential for chondrocyte hypertrophy during skeletal development in mice. Development (Cambridge), 2012, 139, 1153-1163.	1.2	77
66	Genetic Mapping of the Camurati-Engelmann Disease Locus to Chromosome 19q13.1-q13.3. American Journal of Human Genetics, 2000, 66, 143-147.	2.6	72
67	Novel SBDS mutations caused by gene conversion in Japanese patients with Shwachman-Diamond syndrome. Human Genetics, 2004, 114, 345-348.	1.8	72
68	TRPV4â€associated skeletal dysplasias. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 190-204.	0.7	71
69	New gene associations in osteoarthritis: what do they provide, and where are we going?. Current Opinion in Rheumatology, 2007, 19, 429-434.	2.0	70
70	Novel and recurrentEBP mutations in X-linked dominant chondrodysplasia punctata. American Journal of Medical Genetics Part A, 2000, 94, 300-305.	2.4	69
71	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
72	Truncating mutations of RB1CC1 in human breast cancer. Nature Genetics, 2002, 31, 285-288.	9.4	67

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73	Expression, Regulation and Function of Asporin, A Susceptibility Gene in Common Bone and Joint Diseases. Current Medicinal Chemistry, 2008, 15, 724-728.	1.2	66
74	SNP rs11190870 near LBX1 is associated with adolescent idiopathic scoliosis in southern Chinese. Journal of Human Genetics, 2012, 57, 244-246.	1.1	64
75	Identification of RB1CC1, a novel human gene that can induce RB1 in various human cells. Oncogene, 2002, 21, 1295-1298.	2.6	62
76	A large-scale genetic association study of ossification of the posterior longitudinal ligament of the spine. Human Genetics, 2006, 119, 611-616.	1.8	62
77	Chondroitin sulfate N-acetylgalactosaminyltransferase-1 is required for normal cartilage development. Biochemical Journal, 2010, 432, 47-55.	1.7	62
78	Nucleotide Pyrophosphatase Gene Polymorphism Associated With Ossification of the Posterior Longitudinal Ligament of the Spine. Journal of Bone and Mineral Research, 2002, 17, 138-144.	3.1	61
79	A novel dominant-negative mutation in Gdf5 generated by ENU mutagenesis impairs joint formation and causes osteoarthritis in mice. Human Molecular Genetics, 2007, 16, 2366-2375.	1.4	61
80	Novel and recurrent mutations clustered in the von Willebrand factor A domain of MATN3 in multiple epiphyseal dysplasia. Human Mutation, 2004, 24, 439-440.	1.1	60
81	Familial Osteoarthritis of the Hip Joint Associated with Acetabular Dysplasia Maps to Chromosome 13q. American Journal of Human Genetics, 2006, 79, 163-168.	2.6	60
82	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
83	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
84	Identification of a Susceptibility Locus for Severe Adolescent Idiopathic Scoliosis on Chromosome 17q24.3. PLoS ONE, 2013, 8, e72802.	1.1	59
85	Association study of COL9A2 with lumbar disc disease in the Japanese population. Journal of Human Genetics, 2006, 51, 1063-1067.	1.1	58
86	Comprehensive genetic analysis of relevant four genes in 49 patients with Marfan syndrome or Marfan-related phenotypes. American Journal of Medical Genetics, Part A, 2006, 140A, 1719-1725.	0.7	57
87	Prevalence of c.1559delT in ALPL, a common mutation resulting in the perinatal (lethal) form of hypophosphatasia in Japanese and effects of the mutation on heterozygous carriers. Journal of Human Genetics, 2011, 56, 166-168.	1.1	57
88	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
89	Mutations of the fibroblast growth factor receptor-3 gene in one familial and six sporadic cases of achondroplasia in Japanese patients. Human Genetics, 1995, 96, 309-11.	1.8	56
90	Novel types of COMP mutations and genotype-phenotype association in pseudoachondroplasia and multiple epiphyseal dysplasia. Human Genetics, 2003, 112, 84-90.	1.8	56

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91	Spondyloâ€epiphyseal dysplasia, Maroteaux type (pseudoâ€Morquio syndrome type 2), and parastremmatic dysplasia are caused by TRPV4 mutations. American Journal of Medical Genetics, Part A, 2010, 152A, 1443-1449.	0.7	56
92	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
93	Molecular pathogenesis of Spondylocheirodysplastic Ehlersâ€Ðanlos syndrome caused by mutant ZIP13 proteins. EMBO Molecular Medicine, 2014, 6, 1028-1042.	3.3	56
94	Carminerin contributes to chondrocyte calcification during endochondral ossification. Nature Medicine, 2006, 12, 665-670.	15.2	55
95	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
96	A functional variant in ZNF512B is associated with susceptibility to amyotrophic lateral sclerosis in Japanese. Human Molecular Genetics, 2011, 20, 3684-3692.	1.4	53
97	Mutations in the N-terminal globular domain of the type X collagen gene (COL10A1) in patients with Schmid metaphyseal chondrodysplasia. Human Mutation, 1997, 9, 131-135.	1.1	52
98	Zonal gene expression of chondrocytes in osteoarthritic cartilage. Arthritis and Rheumatism, 2008, 58, 3843-3853.	6.7	51
99	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
100	Large replication study and meta-analyses of DVWA as an osteoarthritis susceptibility locus in European and Asian populations. Human Molecular Genetics, 2009, 18, 1518-1523.	1.4	50
101	Intrafamilial phenotypic variability in Engelmann disease (ED): Are ED and Ribbing disease the same entity?. , 2000, 91, 153-156.		49
102	Prediction model for knee osteoarthritis based on genetic and clinical information. Arthritis Research and Therapy, 2010, 12, R187.	1.6	49
103	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
104	A multi-ethnic meta-analysis identifies novel genes, including ACSL5, associated with amyotrophic lateral sclerosis. Communications Biology, 2020, 3, 526.	2.0	49
105	Mutation of the Type X Collagen Gene (COL10A1) Causes Spondylometaphyseal Dysplasia. American Journal of Human Genetics, 1998, 63, 1659-1662.	2.6	48
106	Regulatory polymorphisms in EGR2 are associated with susceptibility to systemic lupus erythematosus. Human Molecular Genetics, 2010, 19, 2313-2320.	1.4	48
107	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
108	Genome-wide association study identifies 14 previously unreported susceptibility loci for adolescent idiopathic scoliosis in Japanese. Nature Communications, 2019, 10, 3685.	5.8	47

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109	Identification and characterization of the human long form of Sox5 (L-SOX5) gene. Gene, 2002, 298, 59-68.	1.0	46
110	Expression and Regulation of the Osteoarthritis-associated Protein Asporin. Journal of Biological Chemistry, 2007, 282, 32193-32199.	1.6	46
111	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
112	Shwachmanâ€“Diamond syndrome is associated with low-turnover osteoporosis. Bone, 2007, 41, 965-972.	1.4	45
113	TRPV4-pathy, a novel channelopathy affecting diverse systems. Journal of Human Genetics, 2010, 55, 400-402.	1.1	45
114	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
115	PAPSS2mutations cause autosomal recessive brachyolmia. Journal of Medical Genetics, 2012, 49, 533-538.	1.5	44
116	Association Analysis of Single Nucleotide Polymorphisms in Cartilage-Specific Collagen Genes With Knee and Hip Osteoarthritis in the Japanese Population. Journal of Bone and Mineral Research, 2002, 17, 1290-1296.	3.1	43
117	The ACVR1 617G>A mutation is also recurrent in three Japanese patients with fibrodysplasia ossificans progressiva. Journal of Human Genetics, 2007, 52, 473-475.	1.1	43
118	Association of the Tag SNPs in the Human SKT Gene (KIAA1217) With Lumbar Disc Herniation. Journal of Bone and Mineral Research, 2009, 24, 1537-1543.	3.1	43
119	Identification of biallelicLRRK1mutations in osteosclerotic metaphyseal dysplasia and evidence for locus heterogeneity. Journal of Medical Genetics, 2016, 53, 568-574.	1.5	43
120	Skewed X-chromosome inactivation causes intra-familial phenotypic variation of an EBP mutation in a family with X-linked dominant chondrodysplasia punctata. Human Genetics, 2003, 112, 78-83.	1.8	41
121	SNPs on chromosome 5p15.3 associated with myocardial infarction in Japanese population. Journal of Human Genetics, 2011, 56, 47-51.	1.1	41
122	Compound Heterozygosity for Null Mutations and a Common Hypomorphic Risk Haplotype inTBX6Causes Congenital Scoliosis. Human Mutation, 2017, 38, 317-323.	1.1	41
123	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
124	Mutation frequencies ofEXT1 andEXT2 in 43 Japanese families with hereditary multiple exostoses. American Journal of Medical Genetics Part A, 2001, 99, 59-62.	2.4	40
125	A functional SNP in EDG2 increases susceptibility to knee osteoarthritis in Japanese. Human Molecular Genetics, 2008, 17, 1790-1797.	1.4	40
126	CANT1 mutation is also responsible for Desbuquois dysplasia, type 2 and Kim variant. Journal of Medical Genetics, 2011, 48, 32-37.	1.5	39

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127	Large-scale screening of TARDBP mutation in amyotrophic lateral sclerosis in Japanese. Neurobiology of Aging, 2012, 33, 786-790.	1.5	39
128	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
129	A functional SNP in ITIH3 is associated with susceptibility to myocardial infarction. Journal of Human Genetics, 2007, 52, 220-229.	1.1	38
130	Molecular Classification of Knee Osteoarthritis. Frontiers in Cell and Developmental Biology, 2021, 9, 725568.	1.8	38
131	The gene for mesomelic dysplasia Kantaputra type is mapped to chromosome 2q24-q32. Journal of Human Genetics, 1998, 43, 32-36.	1.1	37
132	Novel deletion mutations of OPTN in amyotrophic lateral sclerosis in Japanese. Neurobiology of Aging, 2012, 33, 1843.e19-1843.e24.	1.5	37
133	Binding characteristics of the osteoarthritis-associated protein asporin. Journal of Bone and Mineral Metabolism, 2010, 28, 395-402.	1.3	35
134	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
135	Optineurin mutations in Japanese amyotrophic lateral sclerosis: Table 1. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 233-235.	0.9	35
136	Genetic study on developmental dysplasia of the hip. European Journal of Clinical Investigation, 2012, 42, 1121-1125.	1.7	35
137	Modeling type II collagenopathy skeletal dysplasia by directed conversion and induced pluripotent stem cells. Human Molecular Genetics, 2015, 24, 299-313.	1.4	35
138	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
139	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
140	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
141	Genetic analysis of skeletal dysplasia: recent advances and perspectives in the post-genome-sequence era. Journal of Human Genetics, 2006, 51, 581-586.	1.1	33
142	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
143	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
144	Identification of a novel LRRK1 mutation in a family with osteosclerotic metaphyseal dysplasia. Journal of Human Genetics, 2017, 62, 437-441.	1.1	33

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145	A multi-ethnic meta-analysis confirms the association of rs6570507 with adolescent idiopathic scoliosis. Scientific Reports, 2018, 8, 11575.	1.6	33
146	Mapping of a gene responsible for twy (tip-toe walking Yoshimura), a mouse model of ossification of the posterior longitudinal ligament of the spine (OPLL). Mammalian Genome, 1998, 9, 155-156.	1.0	32
147	COL2A1â€"related skeletal dysplasias with predominant metaphyseal involvement. American Journal of Medical Genetics, Part A, 2007, 143A, 161-167.	0.7	32
148	A variant of Desbuquois dysplasia characterized by advanced carpal bone age, short metacarpals, and elongated phalanges: Report of seven cases. American Journal of Medical Genetics, Part A, 2010, 152A, 875-885.	0.7	32
149	Eight novel susceptibility loci and putative causal variants in atopic dermatitis. Journal of Allergy and Clinical Immunology, 2021, 148, 1293-1306.	1.5	32
150	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. PLoS ONE, 2016, 11, e0150555.	1.1	32
151	RMRP mutations in Japanese patients with cartilage-hair hypoplasia. American Journal of Medical Genetics Part A, 2003, 123A, 253-256.	2.4	31
152	Association of the aspartic acid-repeat polymorphism in the asporin gene with age at onset of knee osteoarthritis in Han Chinese Population. Journal of Human Genetics, 2007, 52, 664-667.	1.1	31
153	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
154	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
155	Cartilage hair hypoplasia mutations that lead to RMRP promoter inefficiency or RNA transcript instability. American Journal of Medical Genetics, Part A, 2007, 143A, 2675-2681.	0.7	30
156	Meta-analysis identifies aMECOMgene as a novel predisposing factor of osteoporotic fracture. Journal of Medical Genetics, 2013, 50, 212-219.	1.5	30
157	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
158	Camurati-Engelmann disease type II: Progressive diaphyseal dysplasia with striations of the bones. American Journal of Medical Genetics Part A, 2002, 107, 5-11.	2.4	29
159	Pre-B-cell leukemia homeobox 1 (PBX1) shows functional and possible genetic association with bone mineral density variation. Human Molecular Genetics, 2009, 18, 679-687.	1.4	29
160	Clinical and Radiographic Features of the Autosomal Recessive form of Brachyolmia Caused byPAPSS2Mutations. Human Mutation, 2013, 34, 1381-1386.	1.1	29
161	Genetics of Ossification of the Posterior Longitudinal Ligament of the Spine: A Mini Review. Journal of Bone Metabolism, 2014, 21, 127.	0.5	29
162	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

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163	BGN Mutations in X-Linked Spondyloepimetaphyseal Dysplasia. American Journal of Human Genetics, 2016, 98, 1243-1248.	2.6	29
164	Transcriptional regulation of the cartilage intermediate layer protein (CILP) gene. Biochemical and Biophysical Research Communications, 2006, 341, 121-127.	1.0	28
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