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List of Publications by Year
in descending order

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65 papers	2,319 citations	201674 27 h-index	233421 45 g-index
70 all docs	70 docs citations	70 times ranked	3896 citing authors

#	Article	IF	Citations
1	Genetic epidemiology, prevalence, and genotypeâ€“phenotype correlations in the Swedish population with osteogenesis imperfecta. European Journal of Human Genetics, 2015, 23, 1042-1050.	2.8	126
2	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	8.2	116
3	FAM111A Mutations Result in Hypoparathyroidism and Impaired Skeletal Development. American Journal of Human Genetics, 2013, 92, 990-995.	6.2	114
4	Demonstration of Estrogen Receptor-Î² Immunoreactivity in Human Growth Plate Cartilage. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 370-373.	3.6	113
5	Mutations in SPINT2 Cause a Syndromic Form of Congenital Sodium Diarrhea. American Journal of Human Genetics, 2009, 84, 188-196.	6.2	110
6	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. American Journal of Human Genetics, 2014, 95, 285-293.	6.2	110
7	Demonstration of Estrogen Receptor-Â Immunoreactivity in Human Growth Plate Cartilage. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 370-373.	3.6	99
8	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. Genome Medicine, 2019, 11, 68.	8.2	88
9	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. Nature Medicine, 2019, 25, 583-590.	30.7	86
10	Extending the phenotype of BMPER-related skeletal dysplasias to ischiospinal dysostosis. Orphanet Journal of Rare Diseases, 2016, 11, 1.	2.7	70
11	Comprehensive genetic and epigenetic analysis of sporadic meningioma for macro-mutations on 22q and micro-mutations within the NF2 locus. BMC Genomics, 2007, 8, 16.	2.8	67
12	Further delineation of the KBG syndrome phenotype caused by ANKRD11 aberrations. European Journal of Human Genetics, 2015, 23, 1176-1185.	2.8	67
13	Mutations in COL1A1 and COL1A2 and dental aberrations in children and adolescents with osteogenesis imperfecta â€“ A retrospective cohort study. PLoS ONE, 2017, 12, e0176466.	2.5	62
14	Analysis of short stature homeobox-containing gene (SHOX) and auxological phenotype in dyschondrosteosis and isolated Madelung deformity. Human Genetics, 2001, 109, 551-558.	3.8	60
15	Novel and recurrent TRPV4 mutations and their association with distinct phenotypes within the TRPV4 dysplasia family. Journal of Medical Genetics, 2010, 47, 704-709.	3.2	58
16	A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. American Journal of Human Genetics, 2018, 103, 553-567.	6.2	58
17	Different mutations inPDE4Dassociated with developmental disorders with mirror phenotypes. Journal of Medical Genetics, 2014, 51, 45-54.	3.2	57
18	Clinical and molecular characterization of duplications encompassing the human SHOX gene reveal a variable effect on stature. American Journal of Medical Genetics, Part A, 2009, 149A, 1407-1414.	1.2	46

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19	Spondyloocular Syndrome: Novel Mutations in XYLT2 Gene and Expansion of the Phenotypic Spectrum. Journal of Bone and Mineral Research, 2016, 31, 1577-1585.	2.8	43
20	Growth in achondroplasia: Development of height, weight, head circumference, and body mass index in a European cohort. American Journal of Medical Genetics, Part A, 2018, 176, 1723-1734.	1.2	42
21	Absence of GP130 cytokine receptor signaling causes extended StÃ¼ve-Wiedemann syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	41
22	Mutations in short stature homeobox containing gene (SHOX) in dyschondrosteosis but not in hypochondroplasia. Human Genetics, 2000, 107, 145-149.	3.8	37
23	Tooth agenesis in osteogenesis imperfecta related to mutations in the collagen type I genes. Oral Diseases, 2017, 23, 42-49.	3.0	36
24	Decreased fracture rate, pharmacogenetics and BMD response in 79 Swedish children with osteogenesis imperfecta types I, III and IV treated with Pamidronate. Bone, 2016, 87, 11-18.	2.9	33
25	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. PLoS ONE, 2016, 11, e0150555.	2.5	32
26	Analysis of Multiple Families With Single Individuals Affected by Pseudohypoparathyroidism Type Ib (PHP1B) Reveals Only One Novel Maternally Inherited GNAS Deletion. Journal of Bone and Mineral Research, 2016, 31, 796-805.	2.8	31
27	Skeletal ciliopathies: a pattern recognition approach. Japanese Journal of Radiology, 2020, 38, 193-206.	2.4	30
28	A novel 13 base pair insertion in the sonic hedgehog ZRS limb enhancer (ZRS/LMBR1) causes preaxial polydactyly with triphalangeal thumb. Human Mutation, 2012, 33, 1063-1066.	2.5	29
29	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbachâ€“Nishimura skeletal dysplasia due to pathogenic variants in ALG9. European Journal of Human Genetics, 2016, 24, 198-207.	2.8	29
30	Development of body proportions in achondroplasia: Sitting height, leg length, arm span, and foot length. American Journal of Medical Genetics, Part A, 2018, 176, 1819-1829.	1.2	26
31	Estrogens and human growth. Journal of Steroid Biochemistry and Molecular Biology, 2000, 74, 383-386.	2.5	25
32	Axial spondylometaphyseal dysplasia is also caused by NEK1 mutations. Journal of Human Genetics, 2017, 62, 503-506.	2.3	25
33	Short Stature in KBG Syndrome: First Responses to Growth Hormone Treatment. Hormone Research in Paediatrics, 2015, 83, 361-364.	1.8	24
34	A Large Inversion Involving GNAS Exon A/B and All Exons Encoding GsÎ± Is Associated With Autosomal Dominant Pseudohypoparathyroidism Type Ib (PHP1B). Journal of Bone and Mineral Research, 2017, 32, 776-783.	2.8	22
35	Novel KIAA0753 mutations extend the phenotype of skeletal ciliopathies. Scientific Reports, 2017, 7, 15585.	3.3	21
36	Expanding the Clinical Spectrum of Phenotypes Caused by Pathogenic Variants in PLOD2. Journal of Bone and Mineral Research, 2018, 33, 753-760.	2.8	20

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37	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008.	2.3	19
38	Deconvolution of seed and RNA-binding protein crosstalk in RNAi-based functional genomics. Nature Genetics, 2018, 50, 657-661.	21.4	18
39	Identification of three novelFGF16mutations in Xâ€linked recessive fusion of the fourth and fifth metacarpals and possible correlation with heart disease. Molecular Genetics & Genomic Medicine, 2014, 2, 402-411.	1.2	17
40	Mesomelic and rhizomelic short stature: The phenotype of combined Leri-Weill dyschondrosteosis and achondroplasia or hypochondroplasia. American Journal of Medical Genetics Part A, 2003, 116A, 61-65.	2.4	16
41	Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. Human Mutation, 2018, 39, 1456-1467.	2.5	16
42	Cerebrospinal Fluid of Newborn Infants Contains a Deglycosylated Form of the Intermediate Filament Nestin. Pediatric Research, 1996, 40, 809-814.	2.3	15
43	<scp>SLC26A2</scp> disease spectrum in SwedenÂâ€“Âhigh frequency of recessive multiple epiphyseal dysplasia (<scp>rMED</scp>). Clinical Genetics, 2015, 87, 273-278.	2.0	13
44	Novel mutations in the LRP5 gene in patients with Osteoporosisâ€pseudoglioma syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3132-3135.	1.2	13
45	SLC4A2 Deficiency Causes a New Type of Osteopetrosis. Journal of Bone and Mineral Research, 2020, 37, 226-235.	2.8	12
46	Molecular and clinical delineation of the 17q22 microdeletion phenotype. European Journal of Human Genetics, 2013, 21, 1085-1092.	2.8	11
47	Defining the clinical phenotype of Saulâ€“Wilson syndrome. Genetics in Medicine, 2020, 22, 857-866.	2.4	11
48	Autosomal recessive mutations in the <scp>COL2A1</scp> gene cause severe spondyloepiphyseal dysplasia. Clinical Genetics, 2015, 87, 496-498.	2.0	9
49	Asn540Lys mutation in fibroblast growth factor receptor 3 and phenotype in hypochondroplasia. Acta Paediatrica, International Journal of Paediatrics, 2000, 89, 1072-1076.	1.5	9
50	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. Npj Genomic Medicine, 2022, 7, 11.	3.8	7
51	Autosomal dominant brachyolmia in a large Swedish family: Phenotypic spectrum and natural course. American Journal of Medical Genetics, Part A, 2014, 164, 1635-1641.	1.2	6
52	A Case with Bladder Exstrophy and Unbalanced X Chromosome Rearrangement. European Journal of Pediatric Surgery, 2014, 24, 353-359.	1.3	5
53	Radiologic Features of Type II and Type XI Collagenopathies. Radiographics, 2021, 41, 192-209.	3.3	5
54	Lack of GNAS Remethylation During Oogenesis May Be a Cause of Sporadic Pseudohypoparathyroidism Type Ib. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e1610-e1619.	3.6	5

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55	Metacarpophalangeal pattern profile analysis in leriâ€weill dyschondrosteosis. Acta Radiologica, 2005, 46, 200-207.	1.1	4
56	The phenotype range of achondrogenesis 1A. , 2013, 161A, n/a-n/a.		4
57	Autosomal recessive brachyolmia: early radiological findings. Skeletal Radiology, 2016, 45, 1557-1560.	2.0	4
58	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for nonâ€oncologic disorders. American Journal of Medical Genetics, Part A, 2021, 185, 517-527.	1.2	3
59	Pathogenenic variant in the COL2A1 gene is associated with Spondyloepiphyseal dysplasia type Stanescu. American Journal of Medical Genetics, Part A, 2016, 170, 266-269.	1.2	2
60	Genotype-Phenotype Correlation of PLOD2 Skeletal Dysplasias Using Structural Information. Journal of Bone and Mineral Research, 2018, 33, 1377-1378.	2.8	2
61	A novel missense mutation Ile538Val in the fibroblast growth factor receptor 3 in hypochondroplasia. Human Mutation, 1998, 11, 333-333.	2.5	2
62	Extending the phenotype of lethal skeletal dysplasia type al Gazali. American Journal of Medical Genetics, Part A, 2011, 155, 1404-1408.	1.2	1
63	SAT0493â€...Farber Disease: First Natural History Cohort Demonstrates a Broad Clinical Spectrum with Implications for Juvenile Idiopathic Arthritis Patients. Annals of the Rheumatic Diseases, 2015, 74, 838.3-839.	0.9	1
64	Exploring human genetic skeletal disorders provides important insights into skeletogenesis and elucidates basic developmental signaling pathways. EBioMedicine, 2020, 62, 103091.	6.1	0
65	Genetics of Achondroplasia and Hypochondroplasia. , 2004, , 349-359.		0