

Emma L. Duncan

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

74 papers	4,594 citations	30 h-index	67 g-index
75 ext. papers	5,397 ext. citations	7.4 avg, IF	4.37 L-index

#	Paper	IF	Citations
74	Genome-wide meta-analysis identifies 56 bone mineral density loci and reveals 14 loci associated with risk of fracture. <i>Nature Genetics</i> , 2012 , 44, 491-501	36.3	866
73	Genome-wide association study of ankylosing spondylitis identifies non-MHC susceptibility loci. <i>Nature Genetics</i> , 2010 , 42, 123-7	36.3	484
72	Whole-genome sequencing identifies EN1 as a determinant of bone density and fracture. <i>Nature</i> , 2015 , 526, 112-7	50.4	308
71	Genome-wide association study using extreme truncate selection identifies novel genes affecting bone mineral density and fracture risk. <i>PLoS Genetics</i> , 2011 , 7, e1001372	6	199
70	WNT16 influences bone mineral density, cortical bone thickness, bone strength, and osteoporotic fracture risk. <i>PLoS Genetics</i> , 2012 , 8, e1002745	6	192
69	Defects in the IFT-B component IFT172 cause Jeune and Mainzer-Saldino syndromes in humans. <i>American Journal of Human Genetics</i> , 2013 , 93, 915-25	11	155
68	Meta-analysis of genome-wide scans provides evidence for sex- and site-specific regulation of bone mass. <i>Journal of Bone and Mineral Research</i> , 2007 , 22, 173-183	6.3	128
67	Genetic analyses in a sample of individuals with high or low BMD shows association with multiple Wnt pathway genes. <i>Journal of Bone and Mineral Research</i> , 2008 , 23, 499-506	6.3	127
66	Association of ERAP1, but not IL23R, with ankylosing spondylitis in a Han Chinese population. <i>Arthritis and Rheumatism</i> , 2009 , 60, 3263-8		116
65	Loci for regulation of bone mineral density in men and women identified by genome wide linkage scan: the FAMOS study. <i>Human Molecular Genetics</i> , 2005 , 14, 943-51	5.6	115
64	Multistage genome-wide association meta-analyses identified two new loci for bone mineral density. <i>Human Molecular Genetics</i> , 2014 , 23, 1923-33	5.6	113
63	Influence of LRP5 polymorphisms on normal variation in BMD. <i>Journal of Bone and Mineral Research</i> , 2004 , 19, 1619-27	6.3	108
62	Common variants in the region around Osterix are associated with bone mineral density and growth in childhood. <i>Human Molecular Genetics</i> , 2009 , 18, 1510-7	5.6	107
61	Illness duration and symptom profile in symptomatic UK school-aged children tested for SARS-CoV-2. <i>The Lancet Child and Adolescent Health</i> , 2021 , 5, 708-718	14.5	98
60	Short-rib polydactyly and Jeune syndromes are caused by mutations in WDR60. <i>American Journal of Human Genetics</i> , 2013 , 93, 515-23	11	92
59	Improving diagnosis of tumor-induced osteomalacia with Gallium-68 DOTATATE PET/CT. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013 , 98, 687-94	5.6	87
58	Mutations in the gene encoding IFT dynein complex component WDR34 cause Jeune asphyxiating thoracic dystrophy. <i>American Journal of Human Genetics</i> , 2013 , 93, 932-44	11	84

57	Marked increase in proton pump inhibitors use in Australia. <i>Pharmacoepidemiology and Drug Safety</i> , 2010 , 19, 1019-24	2.6	84
56	Site and gender specificity of inheritance of bone mineral density. <i>Journal of Bone and Mineral Research</i> , 2003 , 18, 1531-8	6.3	81
55	Genetic determinants of heel bone properties: genome-wide association meta-analysis and replication in the GEFOS/GENOMOS consortium. <i>Human Molecular Genetics</i> , 2014 , 23, 3054-68	5.6	78
54	A novel serogenetic approach determines the community prevalence of celiac disease and informs improved diagnostic pathways. <i>BMC Medicine</i> , 2013 , 11, 188	11.4	72
53	Multicentric carpotarsal osteolysis is caused by mutations clustering in the amino-terminal transcriptional activation domain of MAFB. <i>American Journal of Human Genetics</i> , 2012 , 90, 494-501	11	71
52	Autosomal dominant spondylocostal dysostosis is caused by mutation in TBX6. <i>Human Molecular Genetics</i> , 2013 , 22, 1625-31	5.6	70
51	Whole-exome re-sequencing in a family quartet identifies POP1 mutations as the cause of a novel skeletal dysplasia. <i>PLoS Genetics</i> , 2011 , 7, e1002027	6	57
50	Qink or swimQan evaluation of the clinical characteristics of individuals with high bone mass. <i>Osteoporosis International</i> , 2012 , 23, 643-54	5.3	43
49	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. <i>American Journal of Human Genetics</i> , 2016 , 99, 392-406	11	34
48	Fryns Syndrome Associated with Recessive Mutations in PIGN in two Separate Families. <i>Human Mutation</i> , 2016 , 37, 695-702	4.7	34
47	Mutations in LTBP3 cause acromicric dysplasia and geleophysic dysplasia. <i>Journal of Medical Genetics</i> , 2016 , 53, 457-64	5.8	34
46	Whole exome sequencing is an efficient and sensitive method for detection of germline mutations in patients with pheochromocytomas and paragangliomas. <i>Clinical Endocrinology</i> , 2014 , 80, 25-33	3.4	32
45	Compound heterozygous mutations in RIPPLY2 associated with vertebral segmentation defects. <i>Human Molecular Genetics</i> , 2015 , 24, 1234-42	5.6	30
44	Whole exome sequencing is an efficient, sensitive and specific method for determining the genetic cause of short-rib thoracic dystrophies. <i>Clinical Genetics</i> , 2015 , 88, 550-7	4	30
43	Clinical review 2: Genetic determinants of bone density and fracture risk--state of the art and future directions. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010 , 95, 2576-87	5.6	30
42	Genome-wide association study for radiographic vertebral fractures: A potential role for the 16q24 BMD locus. <i>Bone</i> , 2014 , 59, 20-27	4.7	29
41	The genetic associations of acute anterior uveitis and their overlap with the genetics of ankylosing spondylitis. <i>Genes and Immunity</i> , 2016 , 17, 46-51	4.4	27
40	Cross-calibration of dual-energy X-ray densitometers for a large, multi-center genetic study of osteoporosis. <i>Osteoporosis International</i> , 2006 , 17, 125-32	5.3	24

39	Analysis of body composition in individuals with high bone mass reveals a marked increase in fat mass in women but not men. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2013 , 98, 818-28	5.6	23
38	The role of 25-hydroxyvitamin D deficiency in promoting insulin resistance and inflammation in patients with chronic kidney disease: a randomised controlled trial. <i>BMC Nephrology</i> , 2009 , 10, 41	2.7	22
37	Identification of IDUA and WNT16 Phosphorylation-Related Non-Synonymous Polymorphisms for Bone Mineral Density in Meta-Analyses of Genome-Wide Association Studies. <i>Journal of Bone and Mineral Research</i> , 2016 , 31, 358-68	6.3	20
36	Identification of a novel FGFR1 MicroRNA target site polymorphism for bone mineral density in meta-analyses of genome-wide association studies. <i>Human Molecular Genetics</i> , 2015 , 24, 4710-27	5.6	19
35	A Rare Mutation in SMAD9 Associated With High Bone Mass Identifies the SMAD-Dependent BMP Signaling Pathway as a Potential Anabolic Target for Osteoporosis. <i>Journal of Bone and Mineral Research</i> , 2020 , 35, 92-105	6.3	19
34	Peripartum management of glycemia in women with type 1 diabetes. <i>Diabetes Care</i> , 2014 , 37, 364-71	14.6	17
33	PTH1R polymorphisms influence BMD variation through effects on the growing skeleton. <i>Calcified Tissue International</i> , 2007 , 81, 270-8	3.9	17
32	Next-generation sequencing: a frameshift in skeletal dysplasia gene discovery. <i>Osteoporosis International</i> , 2014 , 25, 407-22	5.3	16
31	The IFITM5 mutation c.-14C > T results in an elongated transcript expressed in human bone; and causes varying phenotypic severity of osteogenesis imperfecta type V. <i>BMC Musculoskeletal Disorders</i> , 2014 , 15, 107	2.8	15
30	Genetic studies in osteoporosis--the end of the beginning. <i>Arthritis Research and Therapy</i> , 2008 , 10, 214	5.7	15
29	Cost-effectiveness Analysis of Routine Screening Using Massively Parallel Sequencing for Maturity-Onset Diabetes of the Young in a Pediatric Diabetes Cohort: Reduced Health System Costs and Improved Patient Quality of Life. <i>Diabetes Care</i> , 2019 , 42, 69-76	14.6	15
28	Meta-analysis of genome-wide studies identifies MEF2C SNPs associated with bone mineral density at forearm. <i>Journal of Medical Genetics</i> , 2013 , 50, 473-8	5.8	14
27	Whole-exome sequencing for mutation detection in pediatric disorders of insulin secretion: Maturity onset diabetes of the young and congenital hyperinsulinism. <i>Pediatric Diabetes</i> , 2018 , 19, 656-662	3.6	13
26	Multiple Endocrine Tumors Associated with Germline MAX Mutations: Multiple Endocrine Neoplasia Type 5?. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021 , 106, 1163-1182	5.6	13
25	Comprehensive genetic screening: The prevalence of maturity-onset diabetes of the young gene variants in a population-based childhood diabetes cohort. <i>Pediatric Diabetes</i> , 2019 , 20, 57-64	3.6	13
24	Predicted Benign and Synonymous Variants in Cause Primary Adrenal Insufficiency Through Missplicing. <i>Journal of the Endocrine Society</i> , 2019 , 3, 201-221	0.4	13
23	COL1A1 C-propeptide cleavage site mutation causes high bone mass, bone fragility and jaw lesions: a new cause of gnathodiaphyseal dysplasia?. <i>Clinical Genetics</i> , 2015 , 88, 49-55	4	12
22	Osteocyte transcriptome mapping identifies a molecular landscape controlling skeletal homeostasis and susceptibility to skeletal disease. <i>Nature Communications</i> , 2021 , 12, 2444	17.4	12

21	Clinical usefulness of comprehensive genetic screening in maturity onset diabetes of the young (MODY): A novel ABCC8 mutation in a previously screened family. <i>Journal of Diabetes</i> , 2018 , 10, 764-767	3.8	10
20	Secondary prevention of osteoporosis in Australia: analysis of government-dispensed prescription data. <i>Drugs and Aging</i> , 2010 , 27, 255-64	4.7	10
19	Homozygous variant in C21orf2 in a case of Jeune syndrome with severe thoracic involvement: Extending the phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1698-1704	2.5	9
18	Primary amenorrhoea with hypertension: undiagnosed 17 β -hydroxylase deficiency. <i>Medical Journal of Australia</i> , 2013 , 199, 556-8	4	7
17	Mapping genes for osteoporosis--old dogs and new tricks. <i>Bone</i> , 2010 , 46, 1219-25	4.7	6
16	Characteristics of Early Paget Disease in SQSTM1 Mutation Carriers: Baseline Analysis of the ZiPP Study Cohort. <i>Journal of Bone and Mineral Research</i> , 2020 , 35, 1246-1252	6.3	4
15	Pituitary gigantism treated successfully with the growth hormone receptor antagonist, pegvisomant. <i>Internal Medicine Journal</i> , 2013 , 43, 345-7	1.6	3
14	Osteocyte Transcriptome Mapping Identifies a Molecular Landscape Controlling Skeletal Homeostasis and Susceptibility to Skeletal Disease		3
13	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. <i>Human Molecular Genetics</i> , 2020 , 29, 3662-3678	5.6	3
12	PET detectives: Molecular imaging for pheochromocytomas and paragangliomas in the genomics era. <i>Clinical Endocrinology</i> , 2021 , 95, 13-28	3.4	3
11	A KCNK16 mutation causing TALK-1 gain of function is associated with maturity-onset diabetes of the young. <i>JCI Insight</i> , 2021 , 6,	9.9	3
10	Multicentric Carpotarsal Osteolysis Is Caused by Mutations Clustering in the Amino-Terminal Transcriptional Activation Domain of MAFB. <i>American Journal of Human Genetics</i> , 2014 , 94, 643	11	2
9	Is there a role or target value for nutritional vitamin D in chronic kidney disease?. <i>Nephrology</i> , 2017 , 22 Suppl 2, 57-64	2.2	1
8	Genome-wide Association Studies		1
7	Gene Testing in Everyday Clinical Use: Lessons from the Bone Clinic. <i>Journal of the Endocrine Society</i> , 2021 , 5, bvaa200	0.4	1
6	Illness Characteristics of COVID-19 in Children Infected with the SARS-CoV-2 Delta Variant. <i>Children</i> , 2022 , 9, 652	2.8	1
5	Germline ERBB3 mutation in familial non-small-cell lung carcinoma: expanding ErbB3 role in oncogenesis. <i>Human Molecular Genetics</i> , 2021 , 30, 2393-2401	5.6	0
4	Conclusions and future directions: The known unknowns		
	<i>Nephrology</i> , 2017 , 22 Suppl 2, 70-71	2.2	

- 3 Response to Comment on Johnson et al. Cost-effectiveness Analysis of Routine Screening Using Massively Parallel Sequencing for Maturity-Onset Diabetes of the Young in a Pediatric Diabetes Cohort: Reduced Health System Costs and Improved Patient Quality of Life. *Diabetes Care* 2019;42:69-76. *Diabetes Care*, **2019**, 42, e79-e80 14.6
- 2 Towards genomewide association studies in osteoporosis Lessons from early scans. *BoneKEy Osteovision*, **2007**, 4, 363-366
- 1 Counting the cost: estimating the number of deaths among recently released prisoners in Australia. *Medical Journal of Australia*, **2011**, 195, 383 4