Emma L. Duncan

List of Publications by Year in Descending Order

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	Illness Characteristics of COVID-19 in Children Infected with the SARS-CoV-2 Delta Variant. *Children* , **2022**, 9, 652	2.8	1
73	Multiple Endocrine Tumors Associated with Germline MAX Mutations: Multiple Endocrine Neoplasia Type 5?. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, 1163-1182	5.6	13
72	Gene Testing in Everyday Clinical Use: Lessons from the Bone Clinic. *Journal of the Endocrine Society* , **2021**, 5, bvaa200	0.4	1
71	Osteocyte transcriptome mapping identifies a molecular landscape controlling skeletal homeostasis and susceptibility to skeletal disease. *Nature Communications*, **2021**, 12, 2444	17.4	12
70	PET detectives: Molecular imaging for phaeochromocytomas and paragangliomas in the genomics era. *Clinical Endocrinology*, **2021**, 95, 13-28	3.4	3
69	Germline ERBB3 mutation in familial non-small-cell lung carcinoma: expanding ErbB3 role in oncogenesis. *Human Molecular Genetics*, **2021**, 30, 2393-2401	5.6	0
68	A KCNK16 mutation causing TALK-1 gain of function is associated with maturity-onset diabetes of the young. *JCI Insight*, **2021**, 6,	9.9	3
67	Illness duration and symptom profile in symptomatic UK school-aged children tested for SARS-CoV-2. *The Lancet Child and Adolescent Health*, **2021**, 5, 708-718	14.5	98
66	Characteristics of Early Paget Disease in SQSTM1 Mutation Carriers: Baseline Analysis of the ZiPP Study Cohort. *Journal of Bone and Mineral Research*, **2020**, 35, 1246-1252	6.3	4
65	A Rare Mutation in SMAD9 Associated With High Bone Mass Identifies the SMAD-Dependent BMP Signaling Pathway as a Potential Anabolic Target for Osteoporosis. *Journal of Bone and Mineral Research*, **2020**, 35, 92-105	6.3	19
64	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. *Human Molecular Genetics*, **2020**, 29, 3662-3678	5.6	3
63	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	
62	Comprehensive genetic screening: The prevalence of maturity-onset diabetes of the young gene variants in a population-based childhood diabetes cohort. *Pediatric Diabetes*, **2019**, 20, 57-64	3.6	13
61	Predicted Benign and Synonymous Variants in Cause Primary Adrenal Insufficiency Through Missplicing. *Journal of the Endocrine Society*, **2019**, 3, 201-221	0.4	13
60	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	14.6	15
59	Whole-exome sequencing for mutation detection in pediatric disorders of insulin secretion: Maturity onset diabetes of the young and congenital hyperinsulinism. *Pediatric Diabetes*, **2018**, 19, 656-662	3.6	13
58	Clinical usefulness of comprehensive genetic screening in maturity onset diabetes of the young (MODY): A novel ABCC8 mutation in a previously screened family. *Journal of Diabetes*, **2018**, 10, 764-767	3.8	10

57	Homozygous variant in C21orf2 in a case of Jeune syndrome with severe thoracic involvement: Extending the phenotypic spectrum. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1698-1704	2.5	9
56	Is there a role or target value for nutritional vitamin D in chronic kidney disease?. *Nephrology*, **2017**, 22 Suppl 2, 57-64	2.2	1
55	Conclusions and future directions: The known unknowns . *Nephrology*, **2017**, 22 Suppl 2, 70-71	2.2	
54	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. *American Journal of Human Genetics*, **2016**, 99, 392-406	11	34
53	The genetic associations of acute anterior uveitis and their overlap with the genetics of ankylosing spondylitis. *Genes and Immunity*, **2016**, 17, 46-51	4.4	27
52	Identification of IDUA and WNT16 Phosphorylation-Related Non-Synonymous Polymorphisms for Bone Mineral Density in Meta-Analyses of Genome-Wide Association Studies. *Journal of Bone and Mineral Research*, **2016**, 31, 358-68	6.3	20
51	Fryns Syndrome Associated with Recessive Mutations in PIGN in two Separate Families. *Human Mutation*, **2016**, 37, 695-702	4.7	34
50	Mutations in LTBP3 cause acromicric dysplasia and geleophysic dysplasia. *Journal of Medical Genetics*, **2016**, 53, 457-64	5.8	34
49	Identification of a novel FGFRL1 MicroRNA target site polymorphism for bone mineral density in meta-analyses of genome-wide association studies. *Human Molecular Genetics*, **2015**, 24, 4710-27	5.6	19
48	Whole-genome sequencing identifies EN1 as a determinant of bone density and fracture. *Nature*, **2015**, 526, 112-7	50.4	308
47	Compound heterozygous mutations in RIPPLY2 associated with vertebral segmentation defects. *Human Molecular Genetics*, **2015**, 24, 1234-42	5.6	30
46	COL1A1 C-propeptide cleavage site mutation causes high bone mass, bone fragility and jaw lesions: a new cause of gnathodiaphyseal dysplasia?. *Clinical Genetics*, **2015**, 88, 49-55	4	12
45	Whole exome sequencing is an efficient, sensitive and specific method for determining the genetic cause of short-rib thoracic dystrophies. *Clinical Genetics*, **2015**, 88, 550-7	4	30
44	Multistage genome-wide association meta-analyses identified two new loci for bone mineral density. *Human Molecular Genetics*, **2014**, 23, 1923-33	5.6	113
43	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. *BMC Musculoskeletal Disorders*, **2014**, 15, 107	2.8	15
42	Whole exome sequencing is an efficient and sensitive method for detection of germline mutations in patients with phaeochromcytomas and paragangliomas. *Clinical Endocrinology*, **2014**, 80, 25-33	3.4	32
41	Multicentric Carpotarsal Osteolysis Is Caused by Mutations Clustering in the Amino-Terminal Transcriptional Activation Domain of MAFB. *American Journal of Human Genetics*, **2014**, 94, 643	11	2
40	Genetic determinants of heel bone properties: genome-wide association meta-analysis and replication in the GEFOS/GENOMOS consortium. *Human Molecular Genetics*, **2014**, 23, 3054-68	5.6	78

39	Peripartum management of glycemia in women with type 1 diabetes. *Diabetes Care*, **2014**, 37, 364-71	14.6	17
38	Genome-wide association study for radiographic vertebral fractures: A potential role for the 16q24 BMD locus. *Bone*, **2014**, 59, 20-27	4.7	29
37	Next-generation sequencing: a frameshift in skeletal dysplasia gene discovery. *Osteoporosis International*, **2014**, 25, 407-22	5.3	16
36	Short-rib polydactyly and Jeune syndromes are caused by mutations in WDR60. *American Journal of Human Genetics*, **2013**, 93, 515-23	11	92
35	Mutations in the gene encoding IFT dynein complex component WDR34 cause Jeune asphyxiating thoracic dystrophy. *American Journal of Human Genetics*, **2013**, 93, 932-44	11	84
34	A novel serogenetic approach determines the community prevalence of celiac disease and informs improved diagnostic pathways. *BMC Medicine*, **2013**, 11, 188	11.4	72
33	Improving diagnosis of tumor-induced osteomalacia with Gallium-68 DOTATATE PET/CT. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, 687-94	5.6	87
32	Defects in the IFT-B component IFT172 cause Jeune and Mainzer-Saldino syndromes in humans. *American Journal of Human Genetics*, **2013**, 93, 915-25	11	155
31	Analysis of body composition in individuals with high bone mass reveals a marked increase in fat mass in women but not men. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, 818-28	5.6	23
30	Autosomal dominant spondylocostal dysostosis is caused by mutation in TBX6. *Human Molecular Genetics*, **2013**, 22, 1625-31	5.6	70
29	Genome-wide Association Studies **2013**, 93-100		1
28	Pituitary gigantism treated successfully with the growth hormone receptor antagonist, pegvisomant. *Internal Medicine Journal*, **2013**, 43, 345-7	1.6	3
27	Meta-analysis of genome-wide studies identifies MEF2C SNPs associated with bone mineral density at forearm. *Journal of Medical Genetics*, **2013**, 50, 473-8	5.8	14
26	Primary amenorrhoea with hypertension: undiagnosed 17-□hydroxylase deficiency. *Medical Journal of Australia*, **2013**, 199, 556-8	4	7
25	Sink or swimQan evaluation of the clinical characteristics of individuals with high bone mass. *Osteoporosis International*, **2012**, 23, 643-54	5.3	43
24	Genome-wide meta-analysis identifies 56 bone mineral density loci and reveals 14 loci associated with risk of fracture. *Nature Genetics*, **2012**, 44, 491-501	36.3	866
23	Multicentric carpotarsal osteolysis is caused by mutations clustering in the amino-terminal transcriptional activation domain of MAFB. *American Journal of Human Genetics*, **2012**, 90, 494-501	11	71
22	WNT16 influences bone mineral density, cortical bone thickness, bone strength, and osteoporotic fracture risk. *PLoS Genetics*, **2012**, 8, e1002745	6	192

21	Genome-wide association study using extreme truncate selection identifies novel genes affecting bone mineral density and fracture risk. *PLoS Genetics*, **2011**, 7, e1001372	6	199
20	Whole-exome re-sequencing in a family quartet identifies POP1 mutations as the cause of a novel skeletal dysplasia. *PLoS Genetics*, **2011**, 7, e1002027	6	57
19	Counting the cost: estimating the number of deaths among recently released prisoners in Australia. *Medical Journal of Australia*, **2011**, 195, 383	4	
18	Genome-wide association study of ankylosing spondylitis identifies non-MHC susceptibility loci. *Nature Genetics*, **2010**, 42, 123-7	36.3	484
17	Clinical review 2: Genetic determinants of bone density and fracture risk--state of the art and future directions. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, 2576-87	5.6	30
16	Mapping genes for osteoporosis--old dogs and new tricks. *Bone*, **2010**, 46, 1219-25	4.7	6
15	Secondary prevention of osteoporosis in Australia: analysis of government-dispensed prescription data. *Drugs and Aging*, **2010**, 27, 255-64	4.7	10
14	Marked increase in proton pump inhibitors use in Australia. *Pharmacoepidemiology and Drug Safety*, **2010**, 19, 1019-24	2.6	84
13	Common variants in the region around Osterix are associated with bone mineral density and growth in childhood. *Human Molecular Genetics*, **2009**, 18, 1510-7	5.6	107
12	Association of ERAP1, but not IL23R, with ankylosing spondylitis in a Han Chinese population. *Arthritis and Rheumatism*, **2009**, 60, 3263-8		116
11	The role of 25-hydroxyvitamin D deficiency in promoting insulin resistance and inflammation in patients with chronic kidney disease: a randomised controlled trial. *BMC Nephrology*, **2009**, 10, 41	2.7	22
10	Genetic studies in osteoporosis--the end of the beginning. *Arthritis Research and Therapy*, **2008**, 10, 214	5.7	15
9	Genetic analyses in a sample of individuals with high or low BMD shows association with multiple Wnt pathway genes. *Journal of Bone and Mineral Research*, **2008**, 23, 499-506	6.3	127
8	PTHR1 polymorphisms influence BMD variation through effects on the growing skeleton. *Calcified Tissue International*, **2007**, 81, 270-8	3.9	17
7	Meta-analysis of genome-wide scans provides evidence for sex- and site-specific regulation of bone mass. *Journal of Bone and Mineral Research*, **2007**, 22, 173-183	6.3	128
6	Towards genomewide association studies in osteoporosis lessons from early scans. *BoneKEy Osteovision*, **2007**, 4, 363-366		
5	Cross-calibration of dual-energy X-ray densitometers for a large, multi-center genetic study of osteoporosis. *Osteoporosis International*, **2006**, 17, 125-32	5.3	24
4	Loci for regulation of bone mineral density in men and women identified by genome wide linkage scan: the FAMOS study. *Human Molecular Genetics*, **2005**, 14, 943-51	5.6	115

3	Influence of LRP5 polymorphisms on normal variation in BMD. *Journal of Bone and Mineral Research* , **2004**, 19, 1619-27	6.3	108
2	Site and gender specificity of inheritance of bone mineral density. *Journal of Bone and Mineral Research*, **2003**, 18, 1531-8	6.3	81
1	Osteocyte Transcriptome Mapping Identifies a Molecular Landscape Controlling Skeletal Homeostasis and Susceptibility to Skeletal Disease		3