Michael P Whyte

List of Publications by Year
in descending order

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209 papers	17,291 citations	20036 63 h-index	17373 126 g-index
216 all docs	216 docs citations	216 times ranked	11654 citing authors

#	Article	IF	Citations
1	Hypophosphatasia: Vitamin B6 status of affected children and adults. Bone, 2022, 154, 116204.	1.4	9
2	Sustained Efficacy and Safety of Burosumab, a Monoclonal Antibody to FGF23, in Children With X-Linked Hypophosphatemia. Journal of Clinical Endocrinology and Metabolism, 2022, 107, 813-824.	1.8	36
3	Osteoprotegerin deficiency and aneurysm formation: Case report of iliac artery aneurysms in Juvenile Paget's disease. Annals of Vascular Surgery Brief Reports and Innovations, 2022, 2, 100065.	0.1	0
4	Periarticular calcifications containing giant pseudo-crystals of francolite in skeletal fluorosis from 1,1-difluoroethane â€˜huffingâ€™. Bone, 2022, , 116421.	1.4	2
5	Skeletal and extraskeletal disorders of biomineralization. Nature Reviews Endocrinology, 2022, 18, 473-489.	4.3	25
6	Dysosteosclerosis: Clinical and Radiological Evolution Reflecting Genetic Heterogeneity. JBMR Plus, 2022, 6, .	1.3	2
7	Pharmacodynamics of asfotase alfa in adults with pediatric-onset hypophosphatasia. Bone, 2021, 142, 115664.	1.4	15
8	Patient-Reported Outcomes from a Randomized, Active-Controlled, Open-Label, Phase 3 Trial of Burosumab Versus Conventional Therapy in Children with X-Linked Hypophosphatemia. Calcified Tissue International, 2021, 108, 622-633.	1.5	26
9	Coalescing expansile skeletal disease: Delineation of an extraordinary osteopathy involving the IFITM5 mutation of osteogenesis imperfecta type V. Bone, 2021, 145, 115835.	1.4	7
10	Non-endemic skeletal fluorosis: Causes and associated secondary hyperparathyroidism (case report) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	1.4	17
11	Vitamin B6 deficiency with normal plasma levels of pyridoxal 5â€²-phosphate in perinatal hypophosphatasia. Bone, 2021, 150, 116007.	1.4	9
12	Adult hypophosphatasia treated with reduced frequency of teriparatide dosing. Journal of Musculoskeletal Neuronal Interactions, 2021, 21, 584-589.	0.1	1
13	Hypophosphatasia. , 2020, , 1569-1599.		3
14	Bruck syndrome 2 variant lacking congenital contractures and involving a novel compound heterozygous PLOD2 mutation. Bone, 2020, 130, 115047.	1.4	14
15	Xâ€Linked Hypophosphatemia: Uniquely Mild Disease Associated With PHEX 3â€²â€UTR Mutation c.*231A>G (A Retrospective Caseâ€“Control Study). Journal of Bone and Mineral Research, 2020, 35, 920-931.	3.1	12
16	Early-onset Paget's disease of bone in a Mexican family caused by a novel tandem duplication (77dup27) in TNFRSF11A that encodes RANK. Bone, 2020, 133, 115224.	1.4	7
17	Hypophosphatemic osteosclerosis, hyperostosis, and enthesopathy associated with novel homozygous mutations of DMP1 encoding dentin matrix protein 1 and SPP1 encoding osteopontin: The first digenic SIBLING protein osteopathy?. Bone, 2020, 132, 115190.	1.4	14
18	Growth Curves for Children with X-linked Hypophosphatemia. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 3243-3249.	1.8	26

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19	High bone mass from mutation of low-density lipoprotein receptor-related protein 6 (LRP6). Bone, 2020, 141, 115550.	1.4	22
20	Persistent idiopathic hyperphosphatasemia from bone alkaline phosphatase in a healthy boy. Bone, 2020, 138, 115459.	1.4	4
21	The two faces of giant cell tumor of bone. Cancer Letters, 2020, 489, 1-8.	3.2	20
22	Genetics of Skeletal Disorders. Handbook of Experimental Pharmacology, 2020, 262, 325-351.	0.9	3
23	ZNF687 Mutations in an Extended Cohort of Neoplastic Transformations in Paget's Disease of Bone: Implications for Clinical Pathology. Journal of Bone and Mineral Research, 2020, 35, 1974-1980.	3.1	9
24	Hyperphosphatemia with low FGF7 and normal FGF23 and sFRP4 levels in the circulation characterizes pediatric hypophosphatasia. Bone, 2020, 134, 115300.	1.4	10
25	Healing of vitamin D deficiency rickets complicating hypophosphatasia suggests a role beyond circulating mineral sufficiency for vitamin D in musculoskeletal health. Bone, 2020, 136, 115322.	1.4	12
26	Juvenile Paget's Disease From Heterozygous Mutation of SP7 Encoding Osterix (Specificity Protein 7,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.4	12
27	Tumor-Induced Osteomalacia: Treatment Progress Using Burosumab, an Anti-FGF23 Monoclonal Antibody. Journal of Bone and Mineral Research, 2020, 36, 625-626.	3.1	2
28	SAT-384 Identification of Heterozygous LRP5 Mutation and a TGFÎ²-1 Variant of Unknown Significance in a Patient with Hearing Loss, High Bone Mass, and Oropharyngeal Exostoses. Journal of the Endocrine Society, 2020, 4, .	0.1	0
29	Skeletal fluorosis in a resettled refugee from Kakuma refugee camp. Lancet, The, 2019, 393, 223-225.	6.3	9
30	Absence of an osteopetrosis phenotype in IKBKG (NEMO) mutation-positive women: A case-control study. Bone, 2019, 121, 243-254.	1.4	4
31	New explanation for autosomal dominant high bone mass: Mutation of low-density lipoprotein receptor-related protein 6. Bone, 2019, 127, 228-243.	1.4	42
32	Burosumab versus conventional therapy in children with X-linked hypophosphataemia: a randomised, active-controlled, open-label, phase 3 trial. Lancet, The, 2019, 393, 2416-2427.	6.3	229
33	No vascular calcification on cardiac computed tomography spanning asfotase alfa treatment for an elderly woman with hypophosphatasia. Bone, 2019, 122, 231-236.	1.4	11
34	Natural History of Perinatal and Infantile Hypophosphatasia: A Retrospective Study. Journal of Pediatrics, 2019, 209, 116-124.e4.	0.9	39
35	Five-year efficacy and safety of asfotase alfa therapy for adults and adolescents with hypophosphatasia. Bone, 2019, 121, 149-162.	1.4	99
36	Asfotase alfa for infants and young children with hypophosphatasia: 7 year outcomes of a single-arm, open-label, phase 2 extension trial. Lancet Diabetes and Endocrinology,the, 2019, 7, 93-105.	5.5	91

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37	Efficacy and safety of burosumab in children aged 1â€“4 years with X-linked hypophosphataemia: a multicentre, open-label, phase 2 trial. Lancet Diabetes and Endocrinology,the, 2019, 7, 189-199.	5.5	115
38	Genetic approaches to metabolic bone diseases. British Journal of Clinical Pharmacology, 2019, 85, 1147-1160.	1.1	21
39	MON-516 Skeletal Fluorosis from Fluorocarbon Inhalation. Journal of the Endocrine Society, 2019, 3, .	0.1	0
40	OR13-2 Burosumab Resulted in Greater Improvement in Rickets Than Conventional Therapy in Children with X-Linked Hypophosphatemia (XLH). Journal of the Endocrine Society, 2019, 3, .	0.1	0
41	Commentary. Clinical Chemistry, 2018, 64, 643-644.	1.5	1
42	Hypophosphatasia: Biochemical hallmarks validate the expanded pediatric clinical nosology. Bone, 2018, 110, 96-106.	1.4	36
43	Validation of a Novel Scoring System for Changes in Skeletal Manifestations of Hypophosphatasia in Newborns, Infants, and Children: The Radiographic Global Impression of Change Scale. Journal of Bone and Mineral Research, 2018, 33, 868-874.	3.1	33
44	Alkaline Phosphatase: Discovery and Naming of Our Favorite Enzyme. Journal of Bone and Mineral Research, 2018, 33, 362-364.	3.1	74
45	Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). Bone, 2018, 107, 161-171.	1.4	23
46	Burosumab Therapy in Children with X-Linked Hypophosphatemia. New England Journal of Medicine, 2018, 378, 1987-1998.	13.9	339
47	Mendelian Disorders of RANKL/OPG/RANK/NF-Î²B Signaling. , 2018, , 453-468.		3
48	Hypophosphatasia and How Alkaline Phosphatase Promotes Mineralization. , 2018, , 481-505.		15
49	Heritable Renal Phosphate Wasting Disorders. , 2018, , 761-782.		1
50	Unique Variant of NOD2 Pediatric Granulomatous Arthritis With Severe 1,25-Dihydroxyvitamin D-Mediated Hypercalcemia and Generalized Osteosclerosis. Journal of Bone and Mineral Research, 2018, 33, 2071-2080.	3.1	9
51	Approach to the Patient With Metabolic Bone Disease. , 2018, , 887-902.		1
52	Sclerosteosis: Report of type 1 or 2 in three Indian Tamil families and literature review. Bone, 2018, 116, 321-332.	1.4	17
53	Hypophosphatasia: Enzyme Replacement Therapy Brings New Opportunities and New Challenges. Journal of Bone and Mineral Research, 2017, 32, 667-675.	3.1	110
54	Hypophosphatasia: An overview For 2017. Bone, 2017, 102, 15-25.	1.4	155

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55	Melorheostosis: Exome sequencing of an associated dermatosis implicates postzygotic mosaicism of mutated KRAS. Bone, 2017, 101, 145-155.	1.4	37
56	Skeletal Fluorosis Due To Inhalation Abuse of a Difluoroethane-Containing Computer Cleaner. Journal of Bone and Mineral Research, 2017, 32, 188-195.	3.1	25
57	Raine Syndrome (OMIM #259775), Caused By FAM20C Mutation, Is Congenital Sclerosing Osteomalacia With Cerebral Calcification (OMIM 259660). Journal of Bone and Mineral Research, 2017, 32, 757-769.	3.1	34
58	Osteopontin and the dento-osseous pathobiology of X-linked hypophosphatemia. Bone, 2017, 95, 151-161.	1.4	66
59	Idiopathic Acquired Osteosclerosis in a Middle-Aged Woman With Systemic Lupus Erythematosus. Journal of Bone and Mineral Research, 2016, 31, 1774-1782.	3.1	3
60	Auricular ossification: A newly recognized feature of osteoprotegerinâ€deficiency juvenile Paget disease. American Journal of Medical Genetics, Part A, 2016, 170, 978-985.	0.7	11
61	Adult Hypophosphatasia Treated with Teriparatide: Report of 2 Patients and Review of the Literature. Endocrine Practice, 2016, 22, 941-950.	1.1	47
62	Hypophosphatasia: Natural history study of 101 affected children investigated at one research center. Bone, 2016, 93, 125-138.	1.4	54
63	Commentary. Clinical Chemistry, 2016, 62, 688-688.	1.5	0
64	Asfotase Alfa Treatment Improves Survival for Perinatal and Infantile Hypophosphatasia. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 334-342.	1.8	189
65	Alkaline Phosphatase and Hypophosphatasia. Calcified Tissue International, 2016, 98, 398-416.	1.5	280
66	Neonatal High Bone Mass With First Mutation of the NF-ÎºB Complex: Heterozygous De Novo Missense (p.Asp512Ser) RELA (Rela/p65). Journal of Bone and Mineral Research, 2016, 31, 163-172.	3.1	21
67	Hypophosphatasia â€" aetiology, nosology, pathogenesis, diagnosis and treatment. Nature Reviews Endocrinology, 2016, 12, 233-246.	4.3	346
68	Congenital insensitivity to pain: Fracturing without apparent skeletal pathobiology caused by an autosomal dominant, second mutation in SCN11A encoding voltage-gated sodium channel 1.9. Bone, 2016, 84, 289-298.	1.4	58
69	Hereditary Disorders of the Skeleton. , 2016, , 1173-1183.e4.		2
70	Asfotase alfa therapy for children with hypophosphatasia. JCI Insight, 2016, 1, e85971.	2.3	123
71	Response to: A Rapid Skeletal Turnover in Radiographic Mimic of Osteopetrosis Might Be Secondary to Systemic Mastocytosis. Journal of Bone and Mineral Research, 2015, 30, 946-946.	3.1	1
72	Lenz-Majewski Hyperostotic Dwarfism with Hyperphosphoserinuria from a Novel Mutation in PTDSS1 Encoding Phosphatidylserine Synthase 1. Journal of Bone and Mineral Research, 2015, 30, 606-614.	3.1	17

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73	PHEX 3â€²-UTR c.*231A>G Near The Polyadenylation Signal Is a Relatively Common, Mild, American Mutation That Masquerades as Sporadic or X-Linked Recessive Hypophosphatemic Rickets. Journal of Bone and Mineral Research, 2015, 30, 137-143.	3.1	20
74	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nature Genetics, 2015, 47, 717-726.	9.4	310
75	Adaptor protein-2 sigma subunit mutations causing familial hypocalciuric hypercalcaemia type 3 (FHH3) demonstrate genotypeâ€“phenotype correlations, codon bias and dominant-negative effects. Human Molecular Genetics, 2015, 24, 5079-5092.	1.4	69
76	Hypophosphatasia: Validation and expansion of the clinical nosology for children from 25years experience with 173 pediatric patients. Bone, 2015, 75, 229-239.	1.4	199
77	Juvenile Paget's disease with heterozygous duplication within TNFRSF11A encoding RANK. Bone, 2014, 68, 153-161.	1.4	42
78	Calcific Periarthritis as the Only Clinical Manifestation of Hypophosphatasia in Middle-Aged Sisters. Journal of Bone and Mineral Research, 2014, 29, 929-934.	3.1	64
79	Atypical Subtrochanteric and Diaphyseal Femoral Fractures: Second Report of a Task Force of the American Society for Bone and Mineral Research. Journal of Bone and Mineral Research, 2014, 29, 1-23.	3.1	1,424
80	Multicentric carpotarsal osteolysis syndrome is caused by only a few domainâ€specific mutations in MAFB, a negative regulator of RANKLâ€induced osteoclastogenesis. American Journal of Medical Genetics, Part A, 2014, 164, 2287-2293.	0.7	36
81	Rapid Skeletal Turnover in a Radiographic Mimic of Osteopetrosis. Journal of Bone and Mineral Research, 2014, 29, 2601-2609.	3.1	12
82	Panostotic Expansile Bone Disease With Massive Jaw Tumor Formation and a Novel Mutation in the Signal Peptide of RANK. Journal of Bone and Mineral Research, 2014, 29, 911-921.	3.1	18
83	Severe skeletal toxicity from protracted etidronate therapy for generalized arterial calcification of infancy. Journal of Bone and Mineral Research, 2013, 28, 419-430.	3.1	74
84	Acute Severe Hypercalcemia After Traumatic Fractures and Immobilization in Hypophosphatasia Complicated by Chronic Renal Failure. Journal of Clinical Endocrinology and Metabolism, 2013, 98, 4606-4612.	1.8	24
85	Outcome of Perinatal Hypophosphatasia in Manitoba Mennonites: A Retrospective Cohort Analysis. JIMD Reports, 2013, 11, 73-78.	0.7	52
86	Juvenile Paget's disease in an Iranian kindred with vitamin D deficiency and novel homozygous TNFRSF11B mutation. Journal of Bone and Mineral Research, 2013, 28, 1501-1508.	3.1	26
87	Hypophosphatasia. , 2013, , 337-360.		26
88	Mendelian Disorders of RANKL/OPG/RANK Signaling. , 2013, , 309-324.		6
89	Pregnancy-Associated Osteoporosis With a Heterozygous Deactivating LDL Receptor-Related Protein 5 (LRP5) Mutation and a Homozygous Methylenetetrahydrofolate Reductase (MTHFR) Polymorphism. Journal of Bone and Mineral Research, 2013, 29, 922-928.	3.1	24
90	Enzyme-Replacement Therapy in Life-Threatening Hypophosphatasia. New England Journal of Medicine, 2012, 366, 904-913.	13.9	463

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91	Dual-Energy X-Ray Absorptiometry Interpretation: A Simple Equation for Height Correction in Preteenage Children. Journal of Clinical Densitometry, 2012, 15, 267-274.	0.5	15
92	Whole-exome sequencing identifies mutations in the nucleoside transporter gene SLC29A3 in dysosteosclerosis, a form of osteopetrosis. Human Molecular Genetics, 2012, 21, 4904-4909.	1.4	81
93	Hypophosphatasia. , 2012, , 771-794.		22
94	Fibrodysplasia ossificans progressiva: Middle-age onset of heterotopic ossification from a unique missense mutation (c.974Gâ€‰>â€‰C, p.G325A) in ACVR1. Journal of Bone and Mineral Research, 2012, 27, 729-737.	3.1	47
95	â€œAtypical femoral fracturesâ€•during bisphosphonate exposure in adult hypophosphatasia. Journal of Bone and Mineral Research, 2012, 27, 987-994.	3.1	159
96	Enzyme replacement prevents enamel defects in hypophosphatasia mice. Journal of Bone and Mineral Research, 2012, 27, 1722-1734.	3.1	74
97	Dose response of bone-targeted enzyme replacement for murine hypophosphatasia. Bone, 2011, 49, 250-256.	1.4	44
98	COL1 C-propeptide cleavage site mutations cause high bone mass osteogenesis imperfecta. Human Mutation, 2011, 32, 598-609.	1.1	119
99	Camurati-engelmann disease: Unique variant featuring a novel mutation in TGFÎ²1 encoding transforming growth factor beta 1 and a missense change in TNFSF11 encoding RANK ligand. Journal of Bone and Mineral Research, 2011, 26, 920-933.	3.1	39
100	Hypophosphatasia: Nonlethal disease despite skeletal presentation in utero (17 new cases and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 38	3.1	123
101	Skeletal Fluorosis from Brewed Tea. Journal of Clinical Endocrinology and Metabolism, 2011, 96, 2318-2324.	1.8	44
102	Approach to the Patient with Metabolic Bone Disease. , 2011, , 807-822.		1
103	Buschke-Ollendorff Syndrome. Archives of Dermatology, 2010, 146, 63-8.	1.7	30
104	Polycystic Bone Disease. Journal of Bone and Mineral Research, 2010, 15, 373-373.	3.1	1
105	Elevated serum lactate dehydrogenase isoenzymes and aspartate transaminase distinguish Albers-SchÃ¶nberg disease (Chloride Channel 7 Deficiency Osteopetrosis) among the sclerosing bone disorders. Journal of Bone and Mineral Research, 2010, 25, 2515-2526.	3.1	38
106	Dysosteosclerosis presents as an â€œOsteoclast-Poorâ€•form of osteopetrosis: Comprehensive investigation of a 3-year-old girl and literature review. Journal of Bone and Mineral Research, 2010, 25, 2527-2539.	3.1	36
107	Atypical subtrochanteric and diaphyseal femoral fractures: Report of a task force of the american society for bone and mineral Research. Journal of Bone and Mineral Research, 2010, 25, 2267-2294.	3.1	994
108	Physiological role of alkaline phosphatase explored in hypophosphatasia. Annals of the New York Academy of Sciences, 2010, 1192, 190-200.	1.8	294

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109	2009 Santa Fe Bone Symposium. Journal of Clinical Densitometry, 2010, 13, 1-9.	0.5	17
110	Hereditary Disorders of the Skeleton. , 2010, , 1250-1260.		0
111	Atypical Femoral Fractures, Bisphosphonates, and Adult Hypophosphatasia. Journal of Bone and Mineral Research, 2009, 24, 1132-1134.	3.1	124
112	Chronic Recurrent Multifocal Osteomyelitis Mimicked in Childhood Hypophosphatasia. Journal of Bone and Mineral Research, 2009, 24, 1493-1505.	3.1	61
113	Enzyme Replacement Therapy for Murine Hypophosphatasia. Journal of Bone and Mineral Research, 2008, 23, 777-787.	3.1	222
114	Skeletal Fluorosis From Instant Tea. Journal of Bone and Mineral Research, 2008, 23, 759-769.	3.1	43
115	Bisphosphonate-Induced Osteopetrosis: Novel Bone Modeling Defects, Metaphyseal Osteopenia, and Osteosclerosis Fractures After Drug Exposure Ceases. Journal of Bone and Mineral Research, 2008, 23, 1698-1707.	3.1	88
116	Autosomal Recessive Hypophosphatasia Manifestingin Uterowith Long Bone Deformity but Showing Spontaneous Postnatal Improvement. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3443-3448.	1.8	28
117	Hypophosphatasia. , 2008, , 1573-1598.		25
118	Infantile Hypophosphatasia: Transplantation Therapy Trial Using Bone Fragments and Cultured Osteoblasts. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 2923-2930.	1.8	108
119	Sporadic Hyperphosphatasia Syndrome Featuring Periostitis and Accelerated Skeletal Turnover without Receptor Activator of Nuclear Factor-Î¹B, Osteoprotegerin, or Sequestosome-1 Gene Defects. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 1897-1901.	1.8	4
120	Adult Hypophosphatasia Treated with Teriparatide. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 1203-1208.	1.8	149
121	Pyridoxine-responsive seizures as the first symptom of infantile hypophosphatasia caused by two novel missense mutations (c.677T>C, p.M226T; c.1112C>T, p.T371I) of the tissue-nonspecific alkaline phosphatase gene. Bone, 2007, 40, 1655-1661.	1.4	141
122	Juvenile Paget's Disease: The Second Reported, Oldest Patient Is Homozygous for the TNFRSF11B â€œBalkanâ€•Mutation (966_969delTGACinsCTT), Which Elevates Circulating Immunoreactive Osteoprotegerin Levels. Journal of Bone and Mineral Research, 2007, 22, 938-946.	3.1	36
123	Recovery From Skeletal Fluorosis. Journal of Bone and Mineral Research, 2007, 22, 1476-1476.	3.1	1
124	Paget's Disease of Bone. New England Journal of Medicine, 2006, 355, 593-600.	13.9	126
125	Fluoride Levels in Bottled Teas. American Journal of Medicine, 2006, 119, 189-190.	0.6	7
126	Homozygosity for TNSALP mutation 1348c>T (Arg433Cys) causes infantile hypophosphatasia manifesting transient disease correction and variably lethal outcome in a kindred of black ancestry. Journal of Pediatrics, 2006, 148, 753-758.	0.9	36

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127	Low Serum Alkaline Phosphatase Activity and Pathologic Fracture: Case Report and Brief Review of Hypophosphatasia Diagnosed in Adulthood. Endocrine Practice, 2006, 12, 676-681.	1.1	56
128	Paget's Disease of Bone and Genetic Disorders of RANKL/OPG/RANK/NF-ÂB Signaling. Annals of the New York Academy of Sciences, 2006, 1068, 143-164.	1.8	84
129	Recovery From Skeletal Fluorosis (an Enigmatic, American Case). Journal of Bone and Mineral Research, 2006, 22, 163-170.	3.1	52
130	Deactivating Germline Mutations in LEMD3 Cause Osteopoikilosis and Buschke-Ollendorff Syndrome, but Not Sporadic Melorheostosis. Journal of Bone and Mineral Research, 2006, 22, 243-250.	3.1	74
131	Manifestations in a family with autosomal dominant bone fragility and limb-girdle myopathy. American Journal of Medical Genetics, Part A, 2006, 140A, 322-330.	0.7	9
132	Rare Bone Diseases. , 2006, , 811-829.		0
133	Skeletal changes in epidermal nevus syndrome: Does focal bone disease harbor clues concerning pathogenesis?. American Journal of Medical Genetics, Part A, 2005, 139A, 67-77.	0.7	42
134	Misinterpretation of Osteodensitometry With High Bone Density. Journal of Clinical Densitometry, 2005, 8, 1-6.	0.5	43
135	Skeletal fluorosis and instant tea. American Journal of Medicine, 2005, 118, 78-82.	0.6	73
136	High Bone Mass. , 2005, , 147-150.		1
137	High-Bone-Mass Disease andLRP5. New England Journal of Medicine, 2004, 350, 2096-2099.	13.9	63
138	Inclusion body myopathy associated with Paget disease of bone and frontotemporal dementia is caused by mutant valosin-containing protein. Nature Genetics, 2004, 36, 377-381.	9.4	1,257
139	Oropharyngeal Skeletal Disease Accompanying High Bone Mass and Novel LRP5 Mutation. Journal of Bone and Mineral Research, 2004, 20, 878-885.	3.1	57
140	Heritable disorders of the RANKL/OPG/RANK signaling pathway. Journal of Musculoskeletal Neuronal Interactions, 2004, 4, 254-67.	0.1	37
141	Marrow Cell Transplantation for Infantile Hypophosphatasia. Journal of Bone and Mineral Research, 2003, 18, 624-636.	3.1	155
142	Bisphosphonate-Induced Osteopetrosis. New England Journal of Medicine, 2003, 349, 457-463.	13.9	409
143	Osteoprotegerin Deficiency and Juvenile Paget's Disease. New England Journal of Medicine, 2002, 347, 175-184.	13.9	412
144	Paternally Inherited Inactivating Mutations of theGNAS1Gene in Progressive Osseous Heteroplasia. New England Journal of Medicine, 2002, 346, 99-106.	13.9	284

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145	Familial Expansile Osteolysis (Excessive RANK Effect) in a 5-Generation American Kindred. Medicine (United States), 2002, 81, 101-121.	0.4	39
146	Denaturing Gradient Gel Electrophoresis Analysis of the Tissue Nonspecific Alkaline Phosphatase Isoenzyme Gene in Hypophosphatasia. Molecular Genetics and Metabolism, 2002, 75, 143-153.	0.5	66
147	Elevated plasma 4-pyridoxic acid in renal insufficiency. American Journal of Clinical Nutrition, 2002, 75, 57-64.	2.2	33
148	Expansile Skeletal Hyperphosphatasia Is Caused by a 15-Base Pair Tandem Duplication in TNFRSF11A Encoding RANK and Is Allelic to Familial Expansile Osteolysis. Journal of Bone and Mineral Research, 2002, 17, 26-29.	3.1	163
149	Clinical Delineation and Localization to Chromosome 9p13.3â€“p12 of a Unique Dominant Disorder in Four Families: Hereditary Inclusion Body Myopathy, Paget Disease of Bone, and Frontotemporal Dementia. Molecular Genetics and Metabolism, 2001, 74, 458-475.	0.5	191
150	Preonset Studies of Spondyloepiphyseal Dysplasia Tarda Caused by a Novel 2-Base Pair Deletion in SEDL Encoding Sedlin*. Journal of Bone and Mineral Research, 2001, 16, 2245-2250.	3.1	7
151	Pseudo-(Tumor-Induced) Rickets. Journal of Bone and Mineral Research, 2001, 16, 1564-1571.	3.1	11
152	Historical Vignette: Hypophosphatasia: Molecular Diagnosis of Rathbun's Original Case. Journal of Bone and Mineral Research, 2001, 16, 1724-1727.	3.1	33
153	Mutations in TNFRSF11A, affecting the signal peptide of RANK, cause familial expansile osteolysis. Nature Genetics, 2000, 24, 45-48.	9.4	457
154	Deficiency of the Î±-Subunit of the Stimulatory G Protein and Severe Extraskeletal Ossification. Journal of Bone and Mineral Research, 2000, 15, 2074-2083.	3.1	110
155	Expansile Skeletal Hyperphosphatasia: A New Familial Metabolic Bone Disease. Journal of Bone and Mineral Research, 2000, 15, 2330-2344.	3.1	63
156	â€œCafÃ©-Au-Lait Spotsâ€•Caused by Vitiligo in McCune-Albright Syndrome. Journal of Bone and Mineral Research, 2000, 15, 2521-2523.	3.1	0
157	Alkaline Phosphatase Knock-Out Mice Recapitulate the Metabolic and Skeletal Defects of Infantile Hypophosphatasia. Journal of Bone and Mineral Research, 1999, 14, 2015-2026.	3.1	343
158	Polycystic Bone Disease: A New, Autosomal Dominant Disorder. Journal of Bone and Mineral Research, 1999, 14, 1261-1271.	3.1	6
159	Mild autosomal dominant hypophosphatasia: In utero presentation in two families. , 1999, 86, 410-415.		61
160	Mild hypophosphatasia mimicking severe osteogenesis imperfecta in utero: Bent but not broken. , 1999, 86, 434-438.		70
161	Bilateral radial ray hypoplasia with multiple epiphyseal dysplasia. , 1998, 77, 182-187.		6
162	Skeletal Disorders Characterized By Osteosclerosis Or Hyperostosis. , 1998, , 697-739e.		11

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163	Calcinosis Universalis Complicating Juvenile Dermatomyositis: Resolution During Probenecid Therapy1. Journal of Clinical Endocrinology and Metabolism, 1997, 82, 3536-3542.	1.8	52
164	Extraskeletal Osteoclastomas Responsive to Dexamethasone Treatment in Paget Bone Disease1. Journal of Clinical Endocrinology and Metabolism, 1997, 82, 3826-3834.	1.8	17
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