Outimaija MÃ¤kitie

List of Publications by Year
in descending order

209 papers	7,099 citations	87401 40 h-index	84171 75 g-index
216 all docs	216 docs citations	216 times ranked	9053 citing authors

#	Article	IF	Citations
1	Prenatal maternal and cord blood vitamin D concentrations and negative affectivity in infancy. European Child and Adolescent Psychiatry, 2023, 32, 601-609.	2.8	3
2	Early-Onset Osteoporosis. Calcified Tissue International, 2022, 110, 546-561.	1.5	34
3	Pregnancy Outcome in Women With APECED (APS-1): A Multicenter Study on 43 Females With 83 Pregnancies. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e528-e537.	1.8	7
4	Iron status in early childhood is modified by diet, sex and growth: Secondary analysis of a randomized controlled vitamin D trial. Clinical Nutrition, 2022, 41, 279-287.	2.3	5
5	Targeted Exome Sequencing of Genes Involved in Rare CNVs in Early-Onset Severe Obesity. Frontiers in Genetics, 2022, 13, 839349.	1.1	3
6	Bone mineral density in very low birthweight adultsâ€"A sibling study. Paediatric and Perinatal Epidemiology, 2022, 36, 665-672.	0.8	7
7	Genetic spectrum of prenatally diagnosed skeletal dysplasias in a Finnish patient cohort. Prenatal Diagnosis, 2022, 42, 1525-1537.	1.1	2
8	Mosaic Deletions of Known Genes Explain Skeletal Dysplasias With High and Low Bone Mass. JBMR Plus, 2022, 6, .	1.3	2
9	Novel form of rhizomelic skeletal dysplasia associated with a homozygous variant in GNPNAT1. Journal of Medical Genetics, 2021, 58, 351-356.	1.5	6
10	Biallelic TMEM251 variants in patients with severe skeletal dysplasia and extreme short stature. Human Mutation, 2021, 42, 89-101.	1.1	16
11	Early Detection of Abnormal Growth Associated with Juvenile Acquired Hypothyroidism. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e739-e748.	1.8	1
12	Vitamin D in Head and Neck Cancer: a Systematic Review. Current Oncology Reports, 2021, 23, 5.	1.8	12
13	The Effects of Vitamin D Supplementation During Infancy on Growth During the First 2 Years of Life. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e1140-e1155.	1.8	6
14	A new family with epiphyseal chondrodysplasia type Miura. American Journal of Medical Genetics, Part A, 2021, 185, 112-118.	0.7	3
15	Spondylocarpotarsal synostosis syndrome due to a novel loss of function FLNB variant: a case report. BMC Musculoskeletal Disorders, 2021, 22, 31.	0.8	2
16	Positive airway pressure therapy for obstructive sleep apnea in patients with Osteogenesis imperfecta: a prospective pilot study. BMC Musculoskeletal Disorders, 2021, 22, 61.	0.8	3
17	Prevalence of and factors influencing vitamin D deficiency in paediatric patients diagnosed with cancer at northern latitudes. Acta Paediatrica, International Journal of Paediatrics, 2021, 110, 2252-2258.	0.7	4
18	Pulmonary Follow-Up Imaging in Cartilage-Hair Hypoplasia: a Prospective Cohort Study. Journal of Clinical Immunology, 2021, 41, 1064-1071.	2.0	0

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19	Bone material properties and response to teriparatide in osteoporosis due to WNT1 and PLS3 mutations. Bone, 2021, 146, 115900.	1.4	15
20	New gene discoveries in skeletal diseases with short stature. Endocrine Connections, 2021, 10, R160-R174.	0.8	7
21	SLC26A2-Associated Diastrophic Dysplasia and rMEDâ€"Clinical Features in Affected Finnish Children and Review of the Literature. Genes, 2021, 12, 714.	1.0	9
22	Towards an ICF-based self-report questionnaire for people with skeletal dysplasia to study health, functioning, disability and accessibility. Orphanet Journal of Rare Diseases, 2021, 16, 236.	1.2	1
23	Oligogenic Inheritance of Monoallelic TRIP11, FKBP10, NEK1, TBX5, and NBAS Variants Leading to a Phenotype Similar to Odontochondrodysplasia. Frontiers in Genetics, 2021, 12, 680838.	1.1	6
24	918-P: Predicting the Onset of APECED Diabetes. Diabetes, 2021, 70, 918-P.	0.3	0
25	An<scp>ARHGAP25</scp>variant links aberrant<scp>Rac1</scp>function to earlyâ€onset skeletal fragility. JBMR Plus, 2021, 5, e10509.	1.3	4
26	Phosphate Concentrations and Modifying Factors in Healthy Children From 12 to 24 Months of Age. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 2865-2875.	1.8	4
27	Fecal Bacteria Implicated in Biofilm Production Are Enriched and Associate to Gastrointestinal Symptoms in Patients With APECED â€“ A Pilot Study. Frontiers in Immunology, 2021, 12, 668219.	2.2	6
28	RAB33B and PCNT variants in two Pakistani families with skeletal dysplasia and short stature. BMC Musculoskeletal Disorders, 2021, 22, 630.	0.8	3
29	Women With Chronic Hypoparathyroidism Have Low Risk of Adverse Pregnancy Outcomes. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 3312-3319.	1.8	7
30	Infections and demanding endocrine care contribute to increased mortality in patients with APECED. European Journal of Endocrinology, 2021, 185, K13-K17.	1.9	2
31	Serum and Urinary Osteocalcin in Healthy 7- to 19-Year-Old Finnish Children and Adolescents. Frontiers in Pediatrics, 2021, 9, 610227.	0.9	6
32	Abnormal Bone Tissue Organization and Osteocyte Lacunocanalicular Network in Earlyâ€Onset Osteoporosis Due to <scp>SGMS2</scp> Mutations. JBMR Plus, 2021, 5, e10537.	1.3	7
33	The â€œGEnomics of Musculo Skeletal Traits TranslatiOnal NEtworkâ€: Origins, Rationale, Organization, and Prospects. Frontiers in Endocrinology, 2021, 12, 709815.	1.5	3
34	A Roadmap to Gene Discoveries and Novel Therapies in Monogenic Low and High Bone Mass Disorders. Frontiers in Endocrinology, 2021, 12, 709711.	1.5	13
35	Effect of High-Dose vs Standard-Dose Vitamin D Supplementation on Neurodevelopment of Healthy Term Infants. JAMA Network Open, 2021, 4, e2124493.	2.8	8
36	Patients with autoimmune polyendocrine syndrome type 1 have an increased susceptibility to severe herpesvirus infections. Clinical Immunology, 2021, 231, 108851.	1.4	20

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37	Calvarial doughnut lesions with bone fragility in a French-Canadian family; case report and review of the literature. Bone Reports, 2021, 15, 101121.	0.2	2
38	Long-term Outcome of Kidney Transplantation in 6 Patients With Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED). Transplantation, 2021, Publish Ahead of Print, .	0.5	1
39	Endocrine Disorders and Genital Infections Impair Gynecological Health in APECED (APS-1). Frontiers in Endocrinology, 2021, 12, 784195.	1.5	2
40	A gene-centric approach to biomarker discovery identifies transglutaminase 1 as an epidermal autoantigen. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	4
41	Craniofacial and Craniocervical Features in Cartilage-Hair Hypoplasia: A Radiological Study of 17 Patients and 34 Controls. Frontiers in Endocrinology, 2021, 12, 741548.	1.5	3
42	Increased Burden of Common Risk Alleles in Children With a Significant Fracture History. Journal of Bone and Mineral Research, 2020, 35, 875-882.	3.1	6
43	Outcomes of 42 pregnancies in 14 women with cartilage-hair hypoplasia: a retrospective cohort study. Orphanet Journal of Rare Diseases, 2020, 15, 326.	1.2	2
44	Recommendations for Diagnosis and Treatment of Pseudohypoparathyroidism and Related Disorders: An Updated Practical Tool for Physicians and Patients. Hormone Research in Paediatrics, 2020, 93, 182-196.	0.8	42
45	Exome Sequencing Reveals a Phenotype Modifying Variant in <scp>ZNF528</scp> in Primary Osteoporosis With a <scp>COL1A2</scp> Deletion. Journal of Bone and Mineral Research, 2020, 35, 2381-2392.	3.1	4
46	Peripheral quantitative computed tomography (pQCT) in 12- and 24-month-old children â€“ Practical aspects and descriptive data. Bone, 2020, 141, 115670.	1.4	3
47	The Safety and Efficacy of Live Viral Vaccines in Patients With Cartilage-Hair Hypoplasia. Frontiers in Immunology, 2020, 11, 2020.	2.2	8
48	Functioning and equality according toÂInternational Classification of Functioning, Disability and HealthÂ(ICF) in people with skeletal dysplasia compared to matched control subjects â€“ a cross-sectional survey study. BMC Musculoskeletal Disorders, 2020, 21, 808.	0.8	4
49	Fibroblast growth factor 23 concentrations and modifying factors in children from age 12 to 24Âmonths. Bone, 2020, 141, 115629.	1.4	4
50	Biallelic variants in four genes underlying recessive osteogenesis imperfecta. European Journal of Medical Genetics, 2020, 63, 103954.	0.7	26
51	Unique, Genderâ€Dependent Serum <scp>microRNA</scp> Profile in <scp>PLS3</scp> Geneâ€Related Osteoporosis. Journal of Bone and Mineral Research, 2020, 35, 1962-1973.	3.1	12
52	A novel homozygous missense variant in MATN3 causes spondylo-epimetaphyseal dysplasia Matrilin 3 type in a consanguineous family. European Journal of Medical Genetics, 2020, 63, 103958.	0.7	2
53	Immunodeficiency in cartilageâ€hair hypoplasia: Pathogenesis, clinical course and management. Scandinavian Journal of Immunology, 2020, 92, e12913.	1.3	23
54	Patients With APECED Have Increased Early Mortality Due to Endocrine Causes, Malignancies and infections. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e2207-e2213.	1.8	25

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55	Rare Variants in Genes Linked to Appetite Control and Hypothalamic Development in Early-Onset Severe Obesity. Frontiers in Endocrinology, 2020, 11, 81.	1.5	19
56	Severe Phenotype of APECED (APS1) Increases Risk for Structural Bone Alterations. Frontiers in Endocrinology, 2020, 11, 109.	1.5	11
57	PLS3 Mutations Cause Severe Age and Sex-Related Spinal Pathology. Frontiers in Endocrinology, 2020, 11, 393.	1.5	15
58	The international X-linked hypophosphataemia (XLH) registry (NCT03193476): rationale for and description of an international, observational study. Orphanet Journal of Rare Diseases, 2020, 15, 172.	1.2	21
59	Biomarkers in WNT1 and PLS3 Osteoporosis: Altered Concentrations of DKK1 and FGF23. Journal of Bone and Mineral Research, 2020, 35, 901-912.	3.1	24
60	Vitamin D status in children with leukemia, its predictors, and association with outcome. Pediatric Blood and Cancer, 2020, 67, e28163.	0.8	22
61	GNAS, PDE4D, and PRKAR1A Mutations and GNAS Methylation Changes Are Not a Common Cause of Isolated Early-Onset Severe Obesity Among Finnish Children. Frontiers in Pediatrics, 2020, 8, 145.	0.9	4
62	â€˜Metaphyseal dysplasia without hypotrichosisâ€™ can present with late-onset extraskeletal manifestations. Journal of Medical Genetics, 2020, 57, 18-22.	1.5	7
63	Novel RPL13 Variants and Variable Clinical Expressivity in a Human Ribosomopathy With Spondyloepimetaphyseal Dysplasia. Journal of Bone and Mineral Research, 2020, 36, 283-297.	3.1	12
64	Pubertal development and premature ovarian insufficiency in patients with APECED. European Journal of Endocrinology, 2020, 183, 513-520.	1.9	9
65	SAT-400 Pregnancy Outcome in Women with Hypoparathyroidism:Aswedishpopulation-Based Cohort Study. Journal of the Endocrine Society, 2020, 4, .	0.1	0
66	An Expert Perspective on Phosphate Dysregulation With a Focus on Chronic Hypophosphatemia. Journal of Bone and Mineral Research, 2020, 37, 12-20.	3.1	11
67	Biallelic KIF24 Variants Are Responsible for a Spectrum of Skeletal Disorders Ranging From Lethal Skeletal Ciliopathy to Severe Acromesomelic Dysplasia. Journal of Bone and Mineral Research, 2020, 37, 1642-1652.	3.1	5
68	Collagen X Biomarker (CXM), Linear Growth, and Bone Development in a Vitamin D Intervention Study in Infants. Journal of Bone and Mineral Research, 2020, 37, 1653-1664.	3.1	2
69	A 30-Year Prospective Follow-Up Study Reveals Risk Factors for Early Death in Cartilage-Hair Hypoplasia. Frontiers in Immunology, 2019, 10, 1581.	2.2	25
70	Gonadal Failure Is Common in Long-Term Survivors of Childhood High-Risk Neuroblastoma Treated With High-Dose Chemotherapy and Autologous Stem Cell Rescue. Frontiers in Endocrinology, 2019, 10, 555.	1.5	11
71	The human long non-coding RNA gene RMRP has pleiotropic effects and regulates cell-cycle progression at G2. Scientific Reports, 2019, 9, 13758.	1.6	22
72	Genetic Variation of the Vitamin D Binding Protein Affects Vitamin D Status and Response to Supplementation in Infants. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 5483-5498.	1.8	26

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73	Mesenchymal Cell-Derived Juxtacrine Wnt1 Signaling Regulates Osteoblast Activity and Osteoclast Differentiation. Journal of Bone and Mineral Research, 2019, 34, 1129-1142.	3.1	29
74	A preliminary transcriptome analysis suggests a transitory effect of vitamin D on mitochondrial function in obese young Finnish subjects. Endocrine Connections, 2019, 8, 559-570.	0.8	6
75	The autoimmune targets in IPEX are dominated by gut epithelial proteins. Journal of Allergy and Clinical Immunology, 2019, 144, 327-330.e8.	1.5	11
76	A Novel Homozygous Frameshift Variant in XYLT2 Causes Spondyloocular Syndrome in a Consanguineous Pakistani Family. Frontiers in Genetics, 2019, 10, 144.	1.1	10
77	High-Dose Vitamin D Supplementation Does Not Prevent Allergic Sensitization of Infants. Journal of Pediatrics, 2019, 209, 139-145.e1.	0.9	50
78	FGF23 and its role in X-linked hypophosphatemia-related morbidity. Orphanet Journal of Rare Diseases, 2019, 14, 58.	1.2	158
79	New Insights Into Monogenic Causes of Osteoporosis. Frontiers in Endocrinology, 2019, 10, 70.	1.5	56
80	Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. American Journal of Human Genetics, 2019, 104, 439-453.	2.6	16
81	Genetic variation in GC and CYP2R1 affects 25-hydroxyvitamin D concentration and skeletal parameters: A genome-wide association study in 24-month-old Finnish children. PLoS Genetics, 2019, 15, e1008530.	1.5	14
82	Gynecologic health in cartilageâ€hair hypoplasia: A survey of 26 adult females. American Journal of Medical Genetics, Part A, 2019, 179, 190-195.	0.7	1
83	High Pregnancy, Cord Blood, and Infant Vitamin D Concentrations May Predict Slower Infant Growth. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 397-407.	1.8	15
84	Recessive multiple epiphyseal dysplasia â€“ Clinical characteristics caused by rare compound heterozygous SLC26A2 genotypes. European Journal of Medical Genetics, 2019, 62, 103573.	0.7	10
85	Novel fibronectin mutations and expansion of the phenotype in spondylometaphyseal dysplasia with â€œcorner fracturesâ€. Bone, 2019, 121, 163-171.	1.4	13
86	Novel variants in natriuretic peptide receptor 2 in unrelated patients with acromesomelic dysplasia type Maroteaux. European Journal of Medical Genetics, 2019, 62, 103554.	0.7	12
87	Vitamin D Status in Children With Hemato-Oncological Diseases in Northern Finland. Clinical Pediatrics, 2019, 58, 241-244.	0.4	1
88	Osteoporosis and skeletal dysplasia caused by pathogenic variants in SGMS2. JCI Insight, 2019, 4, .	2.3	47
89	Expansion of the clinical spectrum of frontometaphyseal dysplasia 2 caused by the recurrent mutation p.Pro485Leu in MAP3K7. European Journal of Medical Genetics, 2018, 61, 612-615.	0.7	4
90	Altered MicroRNA Profile in Osteoporosis Caused by Impaired WNT Signaling. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 1985-1996.	1.8	65

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91	Autosomal Recessive Osteogenesis Imperfecta Caused by a Novel Homozygous COL1A2 Mutation. Calcified Tissue International, 2018, 103, 353-358.	1.5	9
92	Prothrombotic state in young females with severe early-onset obesity. Pediatric Research, 2018, 83, 2-4.	1.1	1
93	Cinacalcet Rectifies Hypercalcemia in a Patient With Familial Hypocalciuric Hypercalcemia Type 2 (FHH2) Caused by a Germline Loss-of-Function Gİ±11 Mutation. Journal of Bone and Mineral Research, 2018, 33, 32-41.	3.1	36
94	Autosomal recessive chondrodysplasia with severe short stature caused by a biallelic COL10A1 variant. Journal of Medical Genetics, 2018, 55, 403-407.	1.5	14
95	Gynecologic assessment of 19 adult females with cartilage-hair hypoplasia â€“ high rate of HPV positivity. Orphanet Journal of Rare Diseases, 2018, 13, 207.	1.2	2
96	A Wide Spectrum of Autoimmune Manifestations and Other Symptoms Suggesting Immune Dysregulation in Patients With Cartilage-Hair Hypoplasia. Frontiers in Immunology, 2018, 9, 2468.	2.2	9
97	Novel mutation G324C in WNT1 mapped in a large Pakistani family with severe recessively inherited Osteogenesis Imperfecta. Journal of Biomedical Science, 2018, 25, 82.	2.6	13
98	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	1.5	28
99	Is sleep apnea underdiagnosed in adult patients with osteogenesis imperfecta? â€“a single-center cross-sectional study. Orphanet Journal of Rare Diseases, 2018, 13, 231.	1.2	7
100	Oral Tongue Malignancies in Autoimmune Polyendocrine Syndrome Type 1. Frontiers in Endocrinology, 2018, 9, 463.	1.5	10
101	Effect of Higher vs Standard Dosage of Vitamin D₃Supplementation on Bone Strength and Infection in Healthy Infants. JAMA Pediatrics, 2018, 172, 646.	3.3	59
102	Testicular Function and Bone in Young Men with Severe Childhood-Onset Obesity. Hormone Research in Paediatrics, 2018, 89, 442-449.	0.8	7
103	Comparing osteonecrosis clinical phenotype, timing, and risk factors in children and young adults treated for acute lymphoblastic leukemia. Pediatric Blood and Cancer, 2018, 65, e27300.	0.8	36
104	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. Nature Reviews Endocrinology, 2018, 14, 476-500.	4.3	224
105	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.	0.7	42
106	A novel MYT1L mutation in a patient with severe earlyâ€onset obesity and intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 1972-1975.	0.7	24
107	Rare Copy Number Variants in Array-Based Comparative Genomic Hybridization in Early-Onset Skeletal Fragility. Frontiers in Endocrinology, 2018, 9, 380.	1.5	20
108	Polyostotic Fibrous Dysplasia With and Without McCuneâ€“Albright Syndromeâ€”Clinical Features in a Nordic Pediatric Cohort. Frontiers in Endocrinology, 2018, 9, 96.	1.5	8

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109	Fatigue and disturbances of sleep in patients with osteogenesis imperfecta â€“ a cross-sectional questionnaire study. BMC Musculoskeletal Disorders, 2018, 19, 3.	0.8	20
110	Diversity of Pubertal Development in Cartilage-Hair Hypoplasia; Two Illustrative Cases. Journal of Pediatric and Adolescent Gynecology, 2018, 31, 422-425.	0.3	2
111	A novel frameshift deletion in PLS3 causing severe primary osteoporosis. Journal of Human Genetics, 2018, 63, 923-926.	1.1	20
112	Vitamin D Status in Children with Leukemia. Blood, 2018, 132, 3973-3973.	0.6	2
113	Teriparatide Treatment in Patients withWNT1orPLS3Mutation-Related Early-Onset Osteoporosis - A Pilot Study. Journal of Clinical Endocrinology and Metabolism, 2017, 102, jc.2016-2423.	1.8	24
114	25-hydroxyvitamin D correlates with inflammatory markers in cord blood of healthy newborns. Pediatric Research, 2017, 81, 731-735.	1.1	14
115	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.	3.1	22
116	Cachexia at diagnosis is associated with poor survival in head and neck cancer patients. Acta Oto-Laryngologica, 2017, 137, 778-785.	0.3	71
117	Abnormally High and Heterogeneous Bone Matrix Mineralization After Childhood Solid Organ Transplantation: A Complex Pathology of Low Bone Turnover and Local Defects in Mineralization. Journal of Bone and Mineral Research, 2017, 32, 1116-1125.	3.1	9
118	Decreased telomere length in children with cartilage-hair hypoplasia. Journal of Medical Genetics, 2017, 54, 365-370.	1.5	17
119	Analysis of clinical and immunologic phenotype in a large cohort of children and adults with cartilage-hair hypoplasia. Journal of Allergy and Clinical Immunology, 2017, 140, 612-614.e5.	1.5	29
120	Impaired WNT signaling and the spineâ€"Heterozygous WNT1 mutation causes severe age-related spinal pathology. Bone, 2017, 101, 3-9.	1.4	25
121	CRTAP variants in earlyâ€onset osteoporosis and recurrent fractures. American Journal of Medical Genetics, Part A, 2017, 173, 806-808.	0.7	2
122	Early presentation of osteonecrosis in acute lymphoblastic leukemia: Two children from the Nordic and Baltic cohort. Pediatric Blood and Cancer, 2017, 64, e26624.	0.8	7
123	Copy Number Variants Are Enriched in Individuals With Early-Onset Obesity and Highlight Novel Pathogenic Pathways. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 3029-3039.	1.8	39
124	Osteocyte Protein Expression Is Altered in Low-Turnover Osteoporosis Caused by Mutations in WNT1 and PLS3. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 2340-2348.	1.8	27
125	Towards evidence-based vitamin D supplementation in infants: vitamin D intervention in infants (VIDI) â€" study design and methods of a randomised controlled double-blinded intervention study. BMC Pediatrics, 2017, 17, 91.	0.7	30
126	No Severe Hypercalcemia with Daily Vitamin D3 Supplementation of up to 30 Âµg during the First Year of Life. Hormone Research in Paediatrics, 2017, 88, 147-154.	0.8	12

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127	Novel mutations in the LRP5 gene in patients with Osteoporosisâ€pseudoglioma syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3132-3135.	0.7	13
128	Sex and Iron Modify Fibroblast Growth Factor 23 Concentration in 1-Year-Old Children. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 4526-4533.	1.8	14
129	High bone mass due to novel LRP5 and AMER1 mutations. European Journal of Medical Genetics, 2017, 60, 675-679.	0.7	10
130	Two novel mutations in XYLT2 cause spondyloocular syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3195-3200.	0.7	22
131	Diverse Autoantibody Reactivity in Cartilage-Hair Hypoplasia. Journal of Clinical Immunology, 2017, 37, 508-510.	2.0	8
132	A History of Cowâ€™s Milk Allergy Is Associated with Lower Vitamin D Status in Schoolchildren. Hormone Research in Paediatrics, 2017, 88, 244-250.	0.8	11
133	PLS3 Deletions Lead to Severe Spinal Osteoporosis and Disturbed Bone Matrix Mineralization. Journal of Bone and Mineral Research, 2017, 32, 2394-2404.	3.1	41
134	Recent Discoveries in Monogenic Disorders of Childhood Bone Fragility. Current Osteoporosis Reports, 2017, 15, 303-310.	1.5	16
135	Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. Human Mutation, 2017, 38, 180-192.	1.1	58
136	High prevalence of bronchiectasis in patients with cartilage-hair hypoplasia. Journal of Allergy and Clinical Immunology, 2017, 139, 375-378.	1.5	9
137	Timing of dental development in osteogenesis imperfecta patients with and without bisphosphonate treatment. Bone, 2017, 94, 29-33.	1.4	25
138	Food and Nutrient Intake and Nutrient Sources in 1-Year-Old Infants in Finland: A Cross-Sectional Analysis. Nutrients, 2017, 9, 1309.	1.7	15
139	Metabolic milieu associates with impaired skeletal characteristics in obesity. PLoS ONE, 2017, 12, e0179660.	1.1	11
140	Spondyloocular Syndrome: Novel Mutations in XYLT2 Gene and Expansion of the Phenotypic Spectrum. Journal of Bone and Mineral Research, 2016, 31, 1577-1585.	3.1	43
141	Skeletal Characteristics of WNT1 Osteoporosis in Children and Young Adults. Journal of Bone and Mineral Research, 2016, 31, 1734-1742.	3.1	46
142	Obese young adults exhibit lower total and lower free serum 25â€hydroxycholecalciferol in a randomized vitamin D intervention. Clinical Endocrinology, 2016, 85, 378-385.	1.2	28
143	Osteoporosis and Bone Mass Disorders: From Gene Pathways to Treatments. Trends in Endocrinology and Metabolism, 2016, 27, 262-281.	3.1	108
144	Impaired growth and intracranial calcifications in autosomal dominant hypocalcemia caused by a GNA11 mutation. European Journal of Endocrinology, 2016, 175, 211-218.	1.9	23

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145	Serum 25-Hydroxyvitamin D in Early Childhood Is Nonlinearly Associated with Allergy. International Archives of Allergy and Immunology, 2016, 170, 141-148.	0.9	10
146	Serum parathyroid hormone is related to genetic variation in vitamin D binding protein with respect to total, free, and bioavailable 25-hydroxyvitamin D in middle-aged Caucasians â€“ a cross-sectional study. BMC Nutrition, 2016, 2, .	0.6	8
147	Value of rare low bone mass diseases for osteoporosis genetics. BoneKEy Reports, 2016, 5, 773.	2.7	13
148	Global Consensus Recommendations on Prevention and Management of Nutritional Rickets. Hormone Research in Paediatrics, 2016, 85, 83-106.	0.8	158
149	Global Consensus Recommendations on Prevention and Management of Nutritional Rickets. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 394-415.	1.8	774
150	Fibroblast Growth Factor 23 Concentrations Reflect Sex Differences in Mineral Metabolism and Growth in Early Infancy. Hormone Research in Paediatrics, 2016, 85, 232-241.	0.8	16
151	Functioning and Challenges in Equality and Accessibility Among People with Short Stature. Studies in Health Technology and Informatics, 2016, 229, 402-4.	0.2	0
152	Bone marrow fat unsaturation in young adults is not affected by present or childhood obesity, but increases with age: A pilot study. Metabolism: Clinical and Experimental, 2015, 64, 1574-1581.	1.5	20
153	New Genetic Forms of Childhood-Onset Primary Osteoporosis. Hormone Research in Paediatrics, 2015, 84, 361-369.	0.8	27
154	Cinacalcet Treatment in an Adolescent With Concurrent 22q11.2 Deletion Syndrome and Familial Hypocalciuric Hypercalcemia Type 3 Caused byAP2S1Mutation. Journal of Clinical Endocrinology and Metabolism, 2015, 100, 2515-2518.	1.8	23
155	FGF23 gene variation and its association with phosphate homeostasis and bone mineral density in Finnish children and adolescents. Bone, 2015, 71, 124-130.	1.4	17
156	Increased Body Adiposity and Serum Leptin Concentrations in Very Long-Term Adult Male Survivors of Childhood Acute Lymphoblastic Leukemia. Hormone Research in Paediatrics, 2015, 84, 108-115.	0.8	12
157	Early Protein Intake Is Associated with Body Composition and Resting Energy Expenditure in Young Adults Born with Very Low Birth Weight. Journal of Nutrition, 2015, 145, 2084-2091.	1.3	18
158	Systematic Growth Monitoring for the Early Detection of Celiac Disease in Children. JAMA Pediatrics, 2015, 169, e1525.	3.3	53
159	A Novel Splice Mutation in PLS3 Causes X-linked Early Onset Low-Turnover Osteoporosis. Journal of Bone and Mineral Research, 2015, 30, 510-518.	3.1	66
160	Low Copy Number of the AMY1 Locus Is Associated with Early-Onset Female Obesity in Finland. PLoS ONE, 2015, 10, e0131883.	1.1	70
161	Altered Osteocyte-Specific Protein Expression in Bone after Childhood Solid Organ Transplantation. PLoS ONE, 2015, 10, e0138156.	1.1	16
162	Suppressed Bone Turnover in Obesity: A Link to Energy Metabolism? A Case-Control Study. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 2155-2163.	1.8	59

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