AgnÃ¨s Linglart

List of Publications by Year
in descending order

173 papers	8,109 citations	47 h-index	82 g-index
		47006	58581
192 all docs	192 docs citations	192 times ranked	7517 citing authors

#	Article	IF	Citations
1	Quantitative analysis of lower limb and pelvic deformities in children with X-linked hypophosphatemic rickets. Orthopaedics and Traumatology: Surgery and Research, 2023, 109, 103187.	2.0	7
2	Prevalence of Enthesopathies in Adults With X-linked Hypophosphatemia: Analysis of Risk Factors. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e224-e235.	3.6	14
3	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.	3.6	36
4	Oral health-related quality of life in patients with X-linked hypophosphatemia: a qualitative exploration. Endocrine Connections, 2022, 11, .	1.9	7
5	European expert consensus on practical management of specific aspects of parathyroid disorders in adults and in pregnancy: recommendations of the ESE Educational Program of Parathyroid Disorders (PARAT 2021). European Journal of Endocrinology, 2022, 186, R33-R63.	3.7	73
6	Dental pulp stem cells as a promising model to study imprinting diseases. International Journal of Oral Science, 2022, 14, 19.	8.6	5
7	Imaging patterns in pediatric hypophosphatasia. Pediatric Radiology, 2022, 52, 998-1006.	2.0	3
8	Consensus statement by the French Society of Endocrinology (SFE) and French Society of Pediatric Endocrinology & Diabetology (SFEDP) on diagnosis of Cushing's syndrome. Annales D'Endocrinologie, 2022, 83, 119-141.	1.4	23
9	Interdisciplinary management of FGF23-related phosphate wasting syndromes: a Consensus Statement on the evaluation, diagnosis and care of patients with X-linked hypophosphataemia. Nature Reviews Endocrinology, 2022, 18, 366-384.	9.6	42
10	Skeletal and extraskeletal disorders of biomineralization. Nature Reviews Endocrinology, 2022, 18, 473-489.	9.6	25
11	Clinical lessons learned in constitutional hypopituitarism from two decades of experience in a large international cohort. Clinical Endocrinology, 2021, 94, 277-289.	2.4	22
12	Determinants of Final Height in Patients Born Small for Gestational Age Treated with Recombinant Growth Hormone. Hormone Research in Paediatrics, 2021, 94, 52-62.	1.8	9
13	Renal Hypophosphatemia. , 2021, , 1-29.		0
14	Presenting features and molecular genetics of primary hyperparathyroidism in the paediatric population. European Journal of Endocrinology, 2021, 184, 343-351.	3.7	9
15	A Novel Familial PHP1B Variant With Incomplete Loss of Methylation at GNAS-A/B and Enhanced Methylation at GNAS-AS2. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 2779-2787.	3.6	6
16	Preexisting autoantibodies to type I IFNs underlie critical COVID-19 pneumonia in patients with APS-1. Journal of Experimental Medicine, 2021, 218, .	8.5	185
17	Contribution of imaging to the diagnosis and follow up of X-linked hypophosphatemia. Archives De Pediatrie, 2021, 28, 594-598.	1.0	3
18	X-linked hypophosphatemia, a genetic and treatable cause of rickets!. Archives De Pediatrie, 2021, 28, 587.	1.0	0

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19	Magnetic resonance imaging is a valuable tool to evaluate the therapeutic efficacy of burosumab in children with X-linked hypophosphatemia. European Journal of Endocrinology, 2021, 185, 475-484.	3.7	4
20	X-linked hypophosphatemia and burosumab: Practical clinical points from the French experience. Joint Bone Spine, 2021, 88, 105208.	1.6	14
21	Orthopedic and neurosurgical care of X-linked hypophosphatemia. Archives De Pediatrie, 2021, 28, 599-605.	1.0	4
22	A novel therapeutic strategy for skeletal disorders: Proof of concept of gene therapy for X-linked hypophosphatemia. Science Advances, 2021, 7, eabj5018.	10.3	2
23	Diagnosis, treatment-monitoring and follow-up of children and adolescents with X-linked hypophosphatemia (XLH). Metabolism: Clinical and Experimental, 2020, 103, 153892.	3.4	46
24	Dental and craniofacial features associated with GNAS loss of function mutations. European Journal of Orthodontics, 2020, 42, 525-533.	2.4	7
25	Management of X-linked hypophosphatemia in adults. Metabolism: Clinical and Experimental, 2020, 103, 154049.	3.4	35
26	Development of Enthesopathies and Joint Structural Damage in a Murine Model of X-Linked Hypophosphatemia. Frontiers in Cell and Developmental Biology, 2020, 8, 854.	3.7	14
27	Burden of Illness in Adults With Hypophosphatasia: Data From the Global Hypophosphatasia Patient Registry. Journal of Bone and Mineral Research, 2020, 35, 2171-2178.	2.8	38
28	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.	1.8	42
29	Cytosolic sequestration of the vitamin D receptor as a therapeutic option for vitamin D-induced hypercalcemia. Nature Communications, 2020, 11, 6249.	12.8	14
30	Hyperparathyroidism in Patients With Xâ€Łinked Hypophosphatemia. Journal of Bone and Mineral Research, 2020, 35, 1263-1273.	2.8	31
31	Clinical characteristics of familial hypocalciuric hypercalcaemia type 1: A multicentre study of 77 adult patients. Clinical Endocrinology, 2020, 93, 248-260.	2.4	14
32	Impact of Early Conventional Treatment on Adult Bone and Joints in a Murine Model of X-Linked Hypophosphatemia. Frontiers in Cell and Developmental Biology, 2020, 8, 591417.	3.7	12
33	FGF23 measurement in burosumab-treated patients: an emerging treatment may induce a new analytical interference. Clinical Chemistry and Laboratory Medicine, 2020, 58, e267-e269.	2.3	16
34	Increased prevalence of overweight and obesity in children with X-linked hypophosphatemia. Endocrine Connections, 2020, 9, 144-153.	1.9	30
35	Pseudohypoparathyroidism type 1B (PHP1B), a rare disorder encountered in adolescence. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1475-1479.	0.9	1
36	Targeted Long-Read Sequencing Identifies a Retrotransposon Insertion as a Cause of Altered GNAS Exon A/B Methylation in a Family With Autosomal Dominant Pseudohypoparathyroidism Type 1b (PHP1B). Journal of Bone and Mineral Research, 2020, 37, 1711-1719.	2.8	9

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37	The Lifelong Impact of X-Linked Hypophosphatemia: Results From a Burden of Disease Survey. Journal of the Endocrine Society, 2019, 3, 1321-1334.	0.2	129
38	GHD Diagnostics in Europe and the US: An Audit of National Guidelines and Practice. Hormone Research in Paediatrics, 2019, 92, 150-156.	1.8	31
39	Association of GNAS imprinting defects and deletions of chromosome 2 in two patients: clues explaining phenotypic heterogeneity in pseudohypoparathyroidism type 1B/iPPSD3. Clinical Epigenetics, 2019, 11, 3.	4.1	4
40	Clinical practice recommendations for the diagnosis and management of X-linked hypophosphataemia. Nature Reviews Nephrology, 2019, 15, 435-455.	9.6	318
41	X-linked hypophosphatemia: Management and treatment prospects. Joint Bone Spine, 2019, 86, 731-738.	1.6	37
42	Diagnostic delay is common among patients with hypophosphatasia: initial findings from a longitudinal, prospective, global registry. BMC Musculoskeletal Disorders, 2019, 20, 80.	1.9	69
43	Transcriptional profiling at the DLK1/MEG3 domain explains clinical overlap between imprinting disorders. Science Advances, 2019, 5, eaau9425.	10.3	29
44	Lâ€™hypophosphatÃ©mie liÃ©e Ã lâ€™XÂ: prise en charge et perspectives thÃ©rapeutiques. Revue Du Rhumatisme Monographies, 2019, 86, 55-63.	0.0	1
45	Safety Outcomes During Pediatric GH Therapy: Final Results From the Prospective GeNeSIS Observational Program. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 379-389.	3.6	51
46	High Incidence of Cranial Synostosis and Chiari I Malformation in Children With X-Linked Hypophosphatemic Rickets (XLHR). Journal of Bone and Mineral Research, 2019, 34, 490-496.	2.8	53
47	SAT-259 Natural History of Anthropometric Parametres of Obesity in Children Affected by X-Linked Hypophosphatemia: Longitudinal Obserbational Study. Journal of the Endocrine Society, 2019, 3, .	0.2	2
48	The current landscape of European registries for rare endocrine conditions. European Journal of Endocrinology, 2019, 180, 89-98.	3.7	25
49	SAT-039 GNAS-miRNAs Are Likely Involved in the Phenotype of Patients with Pseudohypoparathyroidism 1B/iPPSD3. Journal of the Endocrine Society, 2019, 3, .	0.2	0
50	SUN-529 Burden of Illness in Adults with Hypophosphatasia: Data From the Global Hypophosphatasia Patient Registry. Journal of the Endocrine Society, 2019, 3, .	0.2	1
51	Genetic and Epigenetic Defects at the GNAS Locus Lead to Distinct Patterns of Skeletal Growth but Similar Early-Onset Obesity. Journal of Bone and Mineral Research, 2018, 33, 1480-1488.	2.8	41
52	15q24.1 BP4-BP1 microdeletion unmasking paternally inherited functional polymorphisms combined with distal 15q24.2q24.3 duplication in a patient with epilepsy, psychomotor delay, overweight, ventricular arrhythmia. European Journal of Medical Genetics, 2018, 61, 459-464.	1.3	11
53	Nutritional management of cow's milk allergy in children: An update. Archives De Pediatrie, 2018, 25, 236-243.	1.0	49
54	Continuous Subcutaneous Recombinant Parathyroid Hormone (1â€“34) Infusion in the Management of Childhood Hypoparathyroidism Associated with Malabsorption. Hormone Research in Paediatrics, 2018, 89, 271-277.	1.8	24

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55	Defective Mineralization in X-Linked Hypophosphatemia Dental Pulp Cell Cultures. Journal of Dental Research, 2018, 97, 184-191.	5.2	22
56	Long-term outcome of liver transplantation in childhood: A study of 20-year survivors. American Journal of Transplantation, 2018, 18, 1680-1689.	4.7	69
57	Parathyroid hormone resistance syndromes â€“ Inactivating PTH/PTHrP signaling disorders (iPPSDs). Best Practice and Research in Clinical Endocrinology and Metabolism, 2018, 32, 941-954.	4.7	15
58	Pseudohypoparathyroidism. Endocrinology and Metabolism Clinics of North America, 2018, 47, 865-888.	3.2	59
59	Burosumab Therapy in Children with X-Linked Hypophosphatemia. New England Journal of Medicine, 2018, 378, 1987-1998.	27.0	339
60	Chromosome 14q32.2 Imprinted Region Disruption as an Alternative Molecular Diagnosis of Silver-Russell Syndrome. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2436-2446.	3.6	48
61	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. Nature Reviews Endocrinology, 2018, 14, 476-500.	9.6	224
62	Hypocalcaemic and hypophosphatemic rickets. Best Practice and Research in Clinical Endocrinology and Metabolism, 2018, 32, 455-476.	4.7	30
63	Impaired mineral quality in dentin in X-linked hypophosphatemia. Connective Tissue Research, 2018, 59, 91-96.	2.3	32
64	Mitotane (opâ€™DDD) restores growth and puberty in nine children with Cushingâ€™s disease. Endocrine Connections, 2018, 7, 1280-1287.	1.9	9
65	Progressive Development of PTH Resistance in Patients With Inactivating Mutations on the Maternal Allele of GNAS. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1844-1850.	3.6	25
66	Bone dysplasia. Annales D'Endocrinologie, 2017, 78, 114-122.	1.4	2
67	Magnetic Resonance Imaging Features as Surrogate Markers of X-Linked Hypophosphatemic Rickets Activity. Hormone Research in Paediatrics, 2017, 87, 244-253.	1.8	22
68	Outcomes of orthopedic surgery in a cohort of 49 patients with X-linked hypophosphatemic rickets (XLHR). Endocrine Connections, 2017, 6, 566-573.	1.9	40
69	Two-year recombinant human growth hormone (rhGH) treatment is more effective in pre-pubertal compared to pubertal short children with X-linked hypophosphatemic rickets (XLHR). Growth Hormone and IGF Research, 2017, 36, 11-15.	1.1	22
70	Monitoring guidance for patients with hypophosphatasia treated with asfotase alfa. Molecular Genetics and Metabolism, 2017, 122, 4-17.	1.1	84
71	Tissue-specific mineralization defects in the periodontium of the Hyp mouse model of X-linked hypophosphatemia. Bone, 2017, 103, 334-346.	2.9	38
72	Phosphate and Vitamin D Prevent Periodontitis in X-Linked Hypophosphatemia. Journal of Dental Research, 2017, 96, 388-395.	5.2	84

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73	Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124.	9.6	336
74	Recent Advances in Imprinting Disorders. Clinical Genetics, 2017, 91, 3-13.	2.0	101
75	Osteopontin and the dento-osseous pathobiology of X-linked hypophosphatemia. Bone, 2017, 95, 151-161.	2.9	66
76	Craniosynostosis and hypophosphatasia. Archives De Pediatrie, 2017, 24, 5S89-5S92.	1.0	14
77	Hypophosphatasia: the contribution of imaging. Archives De Pediatrie, 2017, 24, 5S74-5S79.	1.0	10
78	Hypophosphatasia in children and adolescents: clinical features and treatment. Archives De Pediatrie, 2017, 24, 5S66-5S70.	1.0	6
79	Hypophosphatasia: better knowledge for better careâ€¦. Archives De Pediatrie, 2017, 24, 5S49-5S50.	1.0	0
80	Serum GH concentrations must now be expressed in mass units in Franceâ€¦as in the rest of the world. Annales D'Endocrinologie, 2017, 78, 488-489.	1.4	0
81	Lower incidence of fracture after IV bisphosphonates in girls with Rett syndrome and severe bone fragility. PLoS ONE, 2017, 12, e0186941.	2.5	11
82	The Importance of Networking in Pseudohypoparathyroidism: EuroPHP Network and Patient Support Associations. Pediatric Endocrinology Reviews, 2017, 15, 92-97.	1.2	2
83	French law: what about a reasoned reimbursement of serum vitamin D assays?. Psychologie & Neuropsychiatrie Du Vieillissement, 2016, 14, 377-382.	0.2	7
84	Reimbursment of the serum CTX assay in France: the clinical biology nomenclature is incoherent. Annales De Biologie Clinique, 2016, 74, 381-383.	0.1	0
85	Claudin-16 Deficiency Impairs Tight Junction Function in Ameloblasts, Leading to Abnormal Enamel Formation. Journal of Bone and Mineral Research, 2016, 31, 498-513.	2.8	50
86	Higher methylation of the IGF1 P2 promoter is associated with idiopathic short stature. Clinical Endocrinology, 2016, 84, 216-221.	2.4	14
87	Genome-wide DNA methylation analysis of pseudohypoparathyroidism patients with GNAS imprinting defects. Clinical Epigenetics, 2016, 8, 10.	4.1	53
88	Familial Hypocalciuric Hypercalcemia Types 1 and 3 and Primary Hyperparathyroidism: Similarities and Differences. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2185-2195.	3.6	97
89	Impaired quality of life in adults with X-linked hypophosphatemia and skeletal symptoms. European Journal of Endocrinology, 2016, 174, 325-333.	3.7	119
90	Hypophosphatasia. Current Osteoporosis Reports, 2016, 14, 95-105.	3.6	98

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91	Score de lâ€™os trabÃ©culaireÂ: le point. Revue Du Rhumatisme (Edition Francaise), 2016, 83, 183-188.	0.0	0
92	The Prevalence of GNAS Deficiency-Related Diseases in a Large Cohort of Patients Characterized by the EuroPHP Network. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 3657-3668.	3.6	66
93	From pseudohypoparathyroidism to inactivating PTH/PTHrP signalling disorder (iPPSD), a novel classification proposed by the EuroPHP network. European Journal of Endocrinology, 2016, 175, P1-P17.	3.7	117
94	Topical Sodium Thiosulfate: A Treatment for Calcifications in Hyperphosphatemic Familial Tumoral Calcinosis?. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2810-2815.	3.6	32
95	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
96	Report of two novel mutations in PTHLH associated with brachydactyly type E and literature review. American Journal of Medical Genetics, Part A, 2016, 170, 734-742.	1.2	28
97	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	3.8	71
98	Quantitative computed tomography in pediatric patients. Diagnostic and Interventional Imaging, 2016, 97, 499-502.	3.2	2
99	Very low frequency of germline GPR101 genetic variation and no biallelic defects with AIP in a large cohort of patients with sporadic pituitary adenomas. European Journal of Endocrinology, 2016, 174, 523-530.	3.7	44
100	The Impact of Pediatric Eosinophilic Esophagitis on Bone Metabolism. Journal of Allergy and Clinical Immunology, 2015, 135, AB46.	2.9	4
101	Functional Characterization of PRKAR1A Mutations Reveals a Unique Molecular Mechanism Causing Acrodysostosis but Multiple Mechanisms Causing Carney Complex. Journal of Biological Chemistry, 2015, 290, 27816-27828.	3.4	28
102	A randomized pilot trial of growth hormone with anastrozole versus growth hormone alone, starting at the very end of puberty in adolescents with idiopathic short stature. International Journal of Pediatric Endocrinology (Springer), 2015, 2015, 4.	1.6	24
103	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. Clinical Epigenetics, 2015, 7, 123.	4.1	174
104	THU0551â€…Quality of Life of Adults with X-Linked Hypophosphatemic Rickets. Annals of the Rheumatic Diseases, 2015, 74, 400.1-400.	0.9	1
105	Novel Microdeletions Affecting the GNAS Locus in Pseudohypoparathyroidism: Characterization of the Underlying Mechanisms. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E681-E687.	3.6	23
106	Multiple hormonal resistances: Diagnosis, evaluation and therapy. Annales D'Endocrinologie, 2015, 76, 98-100.	1.4	4
107	Trabecular Bone Score: Where are we now?. Joint Bone Spine, 2015, 82, 320-325.	1.6	45
108	Loss of Methylation at GNAS Exon A/B Is Associated With Increased Intrauterine Growth. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E623-E631.	3.6	28

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109	Growth hormone treatment for childhood short stature and risk of stroke in early adulthood. Neurology, 2015, 84, 1062-1063.	1.1	2
110	Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. Clinical Epigenetics, 2015, 7, 23.	4.1	23
111	Macroprolactinomas in Children and Adolescents: Factors Associated With the Response to Treatment in 77 Patients. Journal of Clinical Endocrinology and Metabolism, 2015, 100, 1177-1186.	3.6	83
112	Genetic and Epigenetic Modulation of Growth Hormone Sensitivity Studied With the IGF-1 Generation Test. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E919-E925.	3.6	14
113	AdolescentÂspinal pain: The pediatric orthopedist's point of view. Orthopaedics and Traumatology: Surgery and Research, 2015, 101, S247-S250.	2.0	18
114	Molecular diagnosis of hypophosphatasia and differential diagnosis by targeted Next Generation Sequencing. Molecular Genetics and Metabolism, 2015, 116, 215-220.	1.1	54
115	Multilocus methylation defects in imprinting disorders. Biomolecular Concepts, 2015, 6, 47-57.	2.2	34
116	Treatment of heterotopic ossifications secondary to pseudohypoparathyroid. Annales D'Endocrinologie, 2015, 76, 183-184.	1.4	5
117	Treatment with rhPTH in children. Annales D'Endocrinologie, 2015, 76, 178-179.	1.4	1
118	European guidance for the molecular diagnosis of pseudohypoparathyroidism not caused by point genetic variants at GNAS: an EQA study. European Journal of Human Genetics, 2015, 23, 438-444.	2.8	27
119	Kidney Function and Influence of Sunlight Exposure in Patients With Impaired 24-Hydroxylation of Vitamin D Due to CYP24A1 Mutations. American Journal of Kidney Diseases, 2015, 65, 122-126.	1.9	67
120	Genetic Testing in Pseudohypoparathyroidism. , 2015, , 373-388.		0
121	Abnormal osteopontin and matrix extracellular phosphoglycoprotein localization, and odontoblast differentiation, in X-linked hypophosphatemic teeth. Connective Tissue Research, 2014, 55, 79-82.	2.3	38
122	Therapeutic management of hypophosphatemic rickets from infancy to adulthood. Endocrine Connections, 2014, 3, R13-R30.	1.9	238
123	Bone, Growth Plate and Mineral Metabolism. Yearbook of Paediatric Endocrinology, 2014, , 63-80.	0.0	0
124	Methylation and Transcripts Expression at the Imprinted GNAS Locus in Human Embryonic and Induced Pluripotent Stem Cells and Their Derivatives. Stem Cell Reports, 2014, 3, 432-443.	4.8	15
125	Analysis of AP2S1, a Calcium-Sensing Receptor Regulator, in Familial and Sporadic Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E469-E473.	3.6	11
126	High Frequency of X Chromosome Abnormalities in Women With Short Stature and Elevated Liver Enzymes. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1592-E1596.	3.6	1

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127	RÃ1e de la vitamine D et risque de maladies auto-immunes/cancers. OCL - Oilseeds and Fats, Crops and Lipids, 2014, 21, D309.	1.4	1
128	Mutations in SLC20A2 are a major cause of familial idiopathic basal ganglia calcification. Neurogenetics, 2013, 14, 11-22.	1.4	131
129	From synthesis to replacement of parathyroid hormone. Lancet Diabetes and Endocrinology,the, 2013, 1, 260-261.	11.4	0
130	<i>GNAS</i>-Related Loss-of-Function Disorders and the Role of Imprinting. Hormone Research in Paediatrics, 2013, 79, 119-129.	1.8	50
131	Simultaneous Hyper- and Hypomethylation at Imprinted Loci in a Subset of Patients withGNASEpimutations Underlies a Complex and Different Mechanism of Multilocus Methylation Defect in Pseudohypoparathyroidism Type 1b. Human Mutation, 2013, 34, 1172-1180.	2.5	43
132	Primary hyperparathyroidism in pregnancy. Endocrine, 2013, 44, 591-597.	2.3	65
133	Clinical utility gene card for: Pseudohypoparathyroidism. European Journal of Human Genetics, 2013, 21, 5-5.	2.8	20
134	MEPE-Derived ASARM Peptide Inhibits Odontogenic Differentiation of Dental Pulp Stem Cells and Impairs Mineralization in Tooth Models of X-Linked Hypophosphatemia. PLoS ONE, 2013, 8, e56749.	2.5	61
135	PRKAR1AandPDE4DMutations Cause Acrodysostosis but Two Distinct Syndromes with or without GPCR-Signaling Hormone Resistance. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E2328-E2338.	3.6	100
136	Acrodysostosis. Hormone and Metabolic Research, 2012, 44, 749-758.	1.5	31
137	Acrodysostosis syndromes. BoneKEy Reports, 2012, 1, 225.	2.7	31
138	De NovoSTX16 Deletions: An Infrequent Cause of Pseudohypoparathyroidism Type Ib that Should Be Excluded in Sporadic Cases. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E2314-E2319.	3.6	32
139	Tooth dentin defects reflect genetic disorders affecting bone mineralization. Bone, 2012, 50, 989-997.	2.9	123
140	Hypoparathyroidism in Children. , 2012, , 299-310.		3
141	Parathormone Resistance in Children. , 2012, , 311-322.		0
142	A Pilot Study of Discontinuous, Insulin-Like Growth Factor 1â€“Dosing Growth Hormone Treatment in Young Children with FGFR3 N540K-Mutated Hypochondroplasia. Journal of Pediatrics, 2012, 160, 849-853.	1.8	10
143	Potent constitutive cyclic AMP-generating activity of XLÎ±s implicates this imprinted GNAS product in the pathogenesis of McCuneâ€“Albright Syndrome and fibrous dysplasia of bone. Bone, 2011, 48, 312-320.	2.9	44
144	Tartrate-resistant acid phosphatase deficiency causes a bone dysplasia with autoimmunity and a type I interferon expression signature. Nature Genetics, 2011, 43, 127-131.	21.4	214

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145	Quantification of the methylation at the GNAS locus identifies subtypes of sporadic pseudohypoparathyroidism type Ib. Journal of Medical Genetics, 2011, 48, 55-63.	3.2	53
146	RecurrentPRKAR1AMutation in Acrodysostosis with Hormone Resistance. New England Journal of Medicine, 2011, 364, 2218-2226.	27.0	162
147	Long-Term Results of Continuous Subcutaneous Recombinant PTH (1-34) Infusion in Children with Refractory Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2011, 96, 3308-3312.	3.6	56
148	Growth hormone treatment before the age of 4 years prevents short stature in young girls with Turner syndrome. European Journal of Endocrinology, 2011, 164, 891-897.	3.7	47
149	Infantile Hypercalcemia and Hypercalciuria: New Insights into a Vitamin D-Dependent Mechanism and Response to Ketoconazole Treatment. Journal of Pediatrics, 2010, 157, 296-302.	1.8	65
150	Resistance to epinephrine and hypersensitivity (hyperresponsiveness) to CB1 antagonists in a patient with pseudohypoparathyroidism type Ic. European Journal of Endocrinology, 2010, 162, 819-824.	3.7	15
151	Near Normalization of Adult Height and Body Proportions by Growth Hormone in Pycnodysostosis. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 2827-2831.	3.6	34
152	Recombinant Human GH Replacement Therapy in Children with Pseudohypoparathyroidism Type Ia: First Study on the Effect on Growth. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 5011-5017.	3.6	55
153	Risk of Corrected QT Interval Prolongation after Pamidronate Infusion in Children. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3768-3770.	3.6	12
154	Gonadotrophic status in adolescents with pituitary stalk interruption syndrome. Clinical Endocrinology, 2008, 69, 105-111.	2.4	32
155	Endocrine Manifestations of the Rapid-Onset Obesity with Hypoventilation, Hypothalamic, Autonomic Dysregulation, and Neural Tumor Syndrome in Childhood. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3971-3980.	3.6	120
156	A Maternal Epimutation of GNAS Leads to Albright Osteodystrophy and Parathyroid Hormone Resistance. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 661-665.	3.6	107
157	Genetic Analysis and Evaluation of Resistance to Thyrotropin and Growth Hormone-Releasing Hormone in Pseudohypoparathyroidism Type Ib. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 3738-3742.	3.6	86
158	Microvascular Diabetes Complications in Wolfram Syndrome (Diabetes Insipidus, Diabetes Mellitus,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	8.6	43
159	Similar clinical and laboratory findings in patients with symptomatic autosomal dominant and sporadic pseudohypoparathyroidism type Ib despite different epigenetic changes at the GNAS locus. Clinical Endocrinology, 2007, 67, 822-831.	2.4	98
160	Autosomal-Dominant Pseudohypoparathyroidism Type Ib is Caused by Different Microdeletions Within or Upstream of the GNAS Locus. Annals of the New York Academy of Sciences, 2006, 1068, 250-255.	3.8	18
161	CodingGNASMutations Leading to Hormone Resistance Impairin VitroAgonist- and Cholera Toxin-Induced Adenosine Cyclic 3â€²,5â€²-Monophosphate Formation Mediated by Human XLÎ±s. Endocrinology, 2006, 147, 2253-2262.	2.8	56
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