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	Burosumab Therapy in Children with X-Linked Hypophosphatemia. New England Journal of Medicine, 2018, 378, 1987-1998.	27.0	339
2	Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124.	9.6	336
3	Deletion of the NESP55 differentially methylated region causes loss of maternal GNAS imprints and pseudohypoparathyroidism type Ib. Nature Genetics, 2005, 37, 25-27.	21.4	321
4	Clinical practice recommendations for the diagnosis and management of X-linked hypophosphataemia. Nature Reviews Nephrology, 2019, 15, 435-455.	9.6	318
5	Therapeutic management of hypophosphatemic rickets from infancy to adulthood. Endocrine Connections, 2014, 3, R13-R30.	1.9	238
6	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. Nature Reviews Endocrinology, 2018, 14, 476-500.	9.6	224
7	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
8	A Novel STX16 Deletion in Autosomal Dominant Pseudohypoparathyroidism Type Ib Redefines the Boundaries of a cis-Acting Imprinting Control Element of GNAS. American Journal of Human Genetics, 2005, 76, 804-814.	6.2	185
9	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
10	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. Clinical Epigenetics, 2015, 7, 123.	4.1	174
11	RecurrentPRKAR1AMutation in Acrodysostosis with Hormone Resistance. New England Journal of Medicine, 2011, 364, 2218-2226.	27.0	162
12	GNAS1Lesions in Pseudohypoparathyroidism Ia and Ic: Genotype Phenotype Relationship and Evidence of the Maternal Transmission of the Hormonal Resistance. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 189-197.	3.6	134
13	Mutations in SLC20A2 are a major cause of familial idiopathic basal ganglia calcification. Neurogenetics, 2013, 14, 11-22.	1.4	131
14	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
15	Tooth dentin defects reflect genetic disorders affecting bone mineralization. Bone, 2012, 50, 989-997.	2.9	123
16	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
17	Impaired quality of life in adults with X-linked hypophosphatemia and skeletal symptoms. European Journal of Endocrinology, 2016, 174, 325-333.	3.7	119
18	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

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19	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
20	Recent Advances in Imprinting Disorders. Clinical Genetics, 2017, 91, 3-13.	2.0	101
21	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
22	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
23	Hypophosphatasia. Current Osteoporosis Reports, 2016, 14, 95-105.	3.6	98
24	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
25	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
26	Monitoring guidance for patients with hypophosphatasia treated with asfotase alfa. Molecular Genetics and Metabolism, 2017, 122, 4-17.	1.1	84
27	Phosphate and Vitamin D Prevent Periodontitis in X-Linked Hypophosphatemia. Journal of Dental Research, 2017, 96, 388-395.	5.2	84
28	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
29	Mechanisms of Ligand Binding to the Parathyroid Hormone (PTH)/PTH-Related Protein Receptor: Selectivity of a Modified PTH(1â€“15) Radioligand for GÎ±S-Coupled Receptor Conformations. Molecular Endocrinology, 2006, 20, 931-943.	3.7	73
30	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
31	Spondyloenchondrodysplasia Due to Mutations in ACP5: A Comprehensive Survey. Journal of Clinical Immunology, 2016, 36, 220-234.	3.8	71
32	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
33	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
34	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
35	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
36	Osteopontin and the dento-osseous pathobiology of X-linked hypophosphatemia. Bone, 2017, 95, 151-161.	2.9	66

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37	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
38	Primary hyperparathyroidism in pregnancy. Endocrine, 2013, 44, 591-597.	2.3	65
39	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
40	Pseudohypoparathyroidism. Endocrinology and Metabolism Clinics of North America, 2018, 47, 865-888.	3.2	59
41	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
42	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
43	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
44	Molecular diagnosis of hypophosphatasia and differential diagnosis by targeted Next Generation Sequencing. Molecular Genetics and Metabolism, 2015, 116, 215-220.	1.1	54
45	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
46	Genome-wide DNA methylation analysis of pseudohypoparathyroidism patients with GNAS imprinting defects. Clinical Epigenetics, 2016, 8, 10.	4.1	53
47	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
48	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
49	<i>GNAS</i>-Related Loss-of-Function Disorders and the Role of Imprinting. Hormone Research in Paediatrics, 2013, 79, 119-129.	1.8	50
50	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
51	Nutritional management of cow's milk allergy in children: An update. Archives De Pediatrie, 2018, 25, 236-243.	1.0	49
52	Molecular Diagnosis of Pseudohypoparathyroidism Type Ib in a Family With Presumed Paroxysmal Dyskinesia. Pediatrics, 2005, 115, e242-e244.	2.1	48
53	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
54	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

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55	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
56	Trabecular Bone Score: Where are we now?. Joint Bone Spine, 2015, 82, 320-325.	1.6	45
57	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
58	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
59	Microvascular Diabetes Complications in Wolfram Syndrome (Diabetes Insipidus, Diabetes Mellitus,) Tj ETQq1 1 0.784314 rgBT /Over	8.6	43
60	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
61	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
62	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
63	Puberty in Subjects with Complete Androgen Insensitivity Syndrome. Hormone Research in Paediatrics, 2006, 65, 126-131.	1.8	41
64	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
65	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
66	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
67	Tissue-specific mineralization defects in the periodontium of the Hyp mouse model of X-linked hypophosphatemia. Bone, 2017, 103, 334-346.	2.9	38
68	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
69	X-linked hypophosphatemia: Management and treatment prospects. Joint Bone Spine, 2019, 86, 731-738.	1.6	37
70	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
71	Management of X-linked hypophosphatemia in adults. Metabolism: Clinical and Experimental, 2020, 103, 154049.	3.4	35
72	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

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73	Multilocus methylation defects in imprinting disorders. Biomolecular Concepts, 2015, 6, 47-57.	2.2	34
74	Gonadotrophic status in adolescents with pituitary stalk interruption syndrome. Clinical Endocrinology, 2008, 69, 105-111.	2.4	32
75	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
76	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
77	Impaired mineral quality in dentin in X-linked hypophosphatemia. Connective Tissue Research, 2018, 59, 91-96.	2.3	32
78	GNAS1 Lesions in Pseudohypoparathyroidism Ia and Ic: Genotype Phenotype Relationship and Evidence of the Maternal Transmission of the Hormonal Resistance. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 189-197.	3.6	32
79	Acrodysostosis. Hormone and Metabolic Research, 2012, 44, 749-758.	1.5	31
80	Acrodysostosis syndromes. BoneKEy Reports, 2012, 1, 225.	2.7	31
81	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
82	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
83	Hyperparathyroidism in Patients With Xâ€Linked Hypophosphatemia. Journal of Bone and Mineral Research, 2020, 35, 1263-1273.	2.8	31
84	Hypocalcaemic and hypophosphatemic rickets. Best Practice and Research in Clinical Endocrinology and Metabolism, 2018, 32, 455-476.	4.7	30
85	Increased prevalence of overweight and obesity in children with X-linked hypophosphatemia. Endocrine Connections, 2020, 9, 144-153.	1.9	30
86	Transcriptional profiling at the DLK1/MEG3 domain explains clinical overlap between imprinting disorders. Science Advances, 2019, 5, eaau9425.	10.3	29
87	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
88	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
89	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
90	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

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91	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
92	The current landscape of European registries for rare endocrine conditions. European Journal of Endocrinology, 2019, 180, 89-98.	3.7	25
93	Skeletal and extraskeletal disorders of biomineralization. Nature Reviews Endocrinology, 2022, 18, 473-489.	9.6	25
94	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
95	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
96	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
97	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
98	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
99	Magnetic Resonance Imaging Features as Surrogate Markers of X-Linked Hypophosphatemic Rickets Activity. Hormone Research in Paediatrics, 2017, 87, 244-253.	1.8	22
100	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
101	Defective Mineralization in X-Linked Hypophosphatemia Dental Pulp Cell Cultures. Journal of Dental Research, 2018, 97, 184-191.	5.2	22
102	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
103	Clinical utility gene card for: Pseudohypoparathyroidism. European Journal of Human Genetics, 2013, 21, 5-5.	2.8	20
104	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
105	AdolescentÂspinal pain: The pediatric orthopedist's point of view. Orthopaedics and Traumatology: Surgery and Research, 2015, 101, S247-S250.	2.0	18
106	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
107	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
108	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

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109	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
110	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
111	Higher methylation of the IGF1 P2 promoter is associated with idiopathic short stature. Clinical Endocrinology, 2016, 84, 216-221.	2.4	14
112	Craniosynostosis and hypophosphatasia. Archives De Pediatrie, 2017, 24, 5S89-5S92.	1.0	14
113	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
114	Cytosolic sequestration of the vitamin D receptor as a therapeutic option for vitamin D-induced hypercalcemia. Nature Communications, 2020, 11, 6249.	12.8	14
115	Clinical characteristics of familial hypocalciuric hypercalcaemia type 1: A multicentre study of 77 adult patients. Clinical Endocrinology, 2020, 93, 248-260.	2.4	14
116	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
117	X-linked hypophosphatemia and burosumab: Practical clinical points from the French experience. Joint Bone Spine, 2021, 88, 105208.	1.6	14
118	Risk of Corrected QT Interval Prolongation after Pamidronate Infusion in Children. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3768-3770.	3.6	12
119	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
120	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
121	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
122	Lower incidence of fracture after IV bisphosphonates in girls with Rett syndrome and severe bone fragility. PLoS ONE, 2017, 12, e0186941.	2.5	11
123	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
124	Hypophosphatasia: the contribution of imaging. Archives De Pediatrie, 2017, 24, 5S74-5S79.	1.0	10
125	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
126	Presenting features and molecular genetics of primary hyperparathyroidism in the paediatric population. European Journal of Endocrinology, 2021, 184, 343-351.	3.7	9

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127	Mitotane (opâ€™DDD) restores growth and puberty in nine children with Cushingâ€™s disease. Endocrine Connections, 2018, 7, 1280-1287.	1.9	9
128	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
129	French law: what about a reasoned reimbursement of serum vitamin D assays?. Psychologie & Neuropsychiatrie Du Vieillissement, 2016, 14, 377-382.	0.2	7
130	Dental and craniofacial features associated with GNAS loss of function mutations. European Journal of Orthodontics, 2020, 42, 525-533.	2.4	7
131	Oral health-related quality of life in patients with X-linked hypophosphatemia: a qualitative exploration. Endocrine Connections, 2022, 11, .	1.9	7
132	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
133	Hypophosphatasia in children and adolescents: clinical features and treatment. Archives De Pediatrie, 2017, 24, 5S66-5S70.	1.0	6
134	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
135	Treatment of heterotopic ossifications secondary to pseudohypoparathyroid. Annales D'Endocrinologie, 2015, 76, 183-184.	1.4	5
136	Dental pulp stem cells as a promising model to study imprinting diseases. International Journal of Oral Science, 2022, 14, 19.	8.6	5
137	The Impact of Pediatric Eosinophilic Esophagitis on Bone Metabolism. Journal of Allergy and Clinical Immunology, 2015, 135, AB46.	2.9	4
138	Multiple hormonal resistances: Diagnosis, evaluation and therapy. Annales D'Endocrinologie, 2015, 76, 98-100.	1.4	4
139	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
140	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
141	Orthopedic and neurosurgical care of X-linked hypophosphatemia. Archives De Pediatrie, 2021, 28, 599-605.	1.0	4
142	Hypoparathyroidism in Children. , 2012, , 299-310.		3
143	Contribution of imaging to the diagnosis and follow up of X-linked hypophosphatemia. Archives De Pediatrie, 2021, 28, 594-598.	1.0	3
144	Imaging patterns in pediatric hypophosphatasia. Pediatric Radiology, 2022, 52, 998-1006.	2.0	3

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145	Growth hormone treatment for childhood short stature and risk of stroke in early adulthood. Neurology, 2015, 84, 1062-1063.	1.1	2
146	Quantitative computed tomography in pediatric patients. Diagnostic and Interventional Imaging, 2016, 97, 499-502.	3.2	2
147	Bone dysplasia. Annales D'Endocrinologie, 2017, 78, 114-122.	1.4	2
148	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
149	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
150	The Importance of Networking in Pseudohypoparathyroidism: EuroPHP Network and Patient Support Associations. Pediatric Endocrinology Reviews, 2017, 15, 92-97.	1.2	2
151	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
152	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
153	THU0551â€…Quality of Life of Adults with X-Linked Hypophosphatemic Rickets. Annals of the Rheumatic Diseases, 2015, 74, 400.1-400.	0.9	1
154	Treatment with rhPTH in children. Annales D'Endocrinologie, 2015, 76, 178-179.	1.4	1
155	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
156	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
157	Pseudohypoparathyroidism type 1B (PHP1B), a rare disorder encountered in adolescence. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1475-1479.	0.9	1
158	X-linked hypophosphatemia, obesity and arterial hypertension: data from the XLH21 study. Pediatric Nephrology, 0, , .	1.7	1
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