Harald Jueppner

List of Publications by Year
in descending order

40 papers	2,600 citations	236925 25 h-index	302126 39 g-index
40 all docs	40 docs citations	40 times ranked	1801 citing authors

#	Article	IF	Citations
1	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
2	Autosomal dominant pseudohypoparathyroidism type Ib is associated with a heterozygous microdeletion that likely disrupts a putative imprinting control element of GNAS. Journal of Clinical Investigation, 2003, 112, 1255-1263.	8.2	226
3	Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. Nature Reviews Endocrinology, 2018, 14, 476-500.	9.6	224
4	Paternal Uniparental Isodisomy of Chromosome 20qâ€"and the Resulting Changes in GNAS1 Methylationâ€"as a Plausible Cause of Pseudohypoparathyroidism. American Journal of Human Genetics, 2001, 68, 1283-1289.	6.2	198
5	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
6	Epigenetic Defects ofGNASin Patients with Pseudohypoparathyroidism and Mild Features of Albrightâ€™s Hereditary Osteodystrophy. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 2370-2373.	3.6	157
7	Phosphate and FGF-23. Kidney International, 2011, 79, S24-S27.	5.2	150
8	FGF-23: More than a regulator of renal phosphate handling?. Journal of Bone and Mineral Research, 2010, 25, 2091-2097.	2.8	141
9	Deletion of the NoncodingGNASAntisense Transcript Causes Pseudohypoparathyroidism Type Ib and Biparental Defects ofGNASMethylationin cis. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3993-4002.	3.6	113
10	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
11	Inherited hypophosphatemic disorders in children and the evolving mechanisms of phosphate regulation. Reviews in Endocrine and Metabolic Disorders, 2008, 9, 171-180.	5.7	78
12	A Homozygous [Cys25]PTH(1-84) Mutation That Impairs PTH/PTHrP Receptor Activation Defines a Novel Form of Hypoparathyroidism. Journal of Bone and Mineral Research, 2015, 30, 1803-1813.	2.8	63
13	Targeted deletion of the Nesp55 DMR defines another Gnas imprinting control region and provides a mouse model of autosomal dominant PHP-Ib. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 9275-9280.	7.1	55
14	PSEUDOHYPOPARATHYROIDISM. Endocrinology and Metabolism Clinics of North America, 2000, 29, 569-589.	3.2	49
15	Molecular Diagnosis of Pseudohypoparathyroidism Type Ib in a Family With Presumed Paroxysmal Dyskinesia. Pediatrics, 2005, 115, e242-e244.	2.1	48
16	Caffey disease: New perspectives on old questions. Bone, 2014, 60, 246-251.	2.9	45
17	Similar frequency of paternal uniparental disomy involving chromosome 20q (patUPD20q) in Japanese and Caucasian patients affected by sporadic pseudohypoparathyroidism type Ib (sporPHP1B). Bone, 2015, 79, 15-20.	2.9	41
18	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

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19	Novel Regulators of Phosphate Homeostasis and Bone Metabolism. Therapeutic Apheresis and Dialysis, 2007, 11, S3-22.	0.9	37
20	Exclusion of the GNAS locus in PHP-Ib patients with broad GNAS methylation changes: Evidence for an autosomal recessive form of PHP-Ib?. Journal of Bone and Mineral Research, 2011, 26, 1854-1863.	2.8	34
21	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
22	Molecular Definition of Pseudohypoparathyroidism Variants. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 1541-1552.	3.6	32
23	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
24	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
25	TSH Elevations as the First Laboratory Evidence for Pseudohypoparathyroidism Type Ib (PHP-Ib). Journal of Bone and Mineral Research, 2015, 30, 906-912.	2.8	28
26	Different Mutations Within or Upstream of the GNAS Locus Cause Distinct Forms of Pseudohypoparathyroidism. Journal of Pediatric Endocrinology and Metabolism, 2006, 19, 641-6.	0.9	25
27	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.	2.8	22
28	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
29	High frequency of paternal iso or heterodisomy at chromosome 20 associated with sporadic pseudohypoparathyroidism 1B. Bone, 2019, 123, 145-152.	2.9	16
30	Î±Klotho: FGF23 coreceptor and FGF23-regulating hormone. Journal of Clinical Investigation, 2012, 122, 4336-4339.	8.2	16
31	A Novel GNAS Duplication Associated With Loss-of-Methylation Restricted to Exon A/B Causes Pseudohypoparathyroidism Type Ib (PHP1B). Journal of Bone and Mineral Research, 2020, 36, 546-552.	2.8	8
32	Pseudohypoparathyroidism type 1B associated with assisted reproductive technology. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 1125-1132.	0.9	7
33	A Heterozygous Splice-Site Mutation in PTHLH Causes Autosomal Dominant Shortening of Metacarpals and Metatarsals. Journal of Bone and Mineral Research, 2019, 34, 482-489.	2.8	7
34	Preferential Maternal Transmission of STX16-GNAS Mutations Responsible for Autosomal Dominant Pseudohypoparathyroidism Type Ib (PHP1B): Another Example of Transmission Ratio Distortion. Journal of Bone and Mineral Research, 2020, 36, 696-703.	2.8	6
35	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
36	Mice maintain predominantly maternal GÎ±s expression throughout life in brown fat tissue (BAT), but not other tissues. Bone, 2017, 103, 177-187.	2.9	5

#	Article	IF	Citations
37	Lack of GNAS Remethylation During Oogenesis May Be a Cause of Sporadic Pseudohypoparathyroidism Type Ib. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e1610-e1619.	3.6	5
38	Progression of PTH Resistance in Autosomal Dominant Pseudohypoparathyroidism Type Ib Due to Maternal STX16 Deletions. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e681-e687.	3.6	3
39	A Distinct Variant of Pseudohypoparathyroidism (<scp>PHP</scp>) First Characterized Some 41â€‰Years Ago Is Caused by the 3â€<scp>kb</scp>STX16 Deletion. JBMR Plus, 2021, 5, e10505.	2.7	1
40	A novel deletion involving the first GNAS exon encoding GsÎ± causes PHP1A without methylation changes at exon A/B. Bone, 2022, 157, 116344.	2.9	0