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List of Publications by Year
in descending order

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59 papers	2,909 citations	172207 29 h-index	174990 52 g-index
66 all docs	66 docs citations	66 times ranked	2259 citing authors

#	Article	IF	Citations
1	Mutations Affecting G-Protein Subunit Î±₁₁in Hypercalcemia and Hypocalcemia. New England Journal of Medicine, 2013, 368, 2476-2486.	13.9	340
2	Mutations in AP2S1 cause familial hypocalciuric hypercalcemia type 3. Nature Genetics, 2013, 45, 93-97.	9.4	242
3	The calcium-sensing receptor in physiology and in calcitropic and noncalcitropic diseases. Nature Reviews Endocrinology, 2019, 15, 33-51.	4.3	226
4	Identification of 70 calcium-sensing receptor mutations in hyper- and hypo-calcaemic patients: evidence for clustering of extracellular domain mutations at calcium-binding sites. Human Molecular Genetics, 2012, 21, 2768-2778.	1.4	154
5	Disorders of the calcium-sensing receptor and partner proteins: insights into the molecular basis of calcium homeostasis. Journal of Molecular Endocrinology, 2016, 57, R127-R142.	1.1	144
6	Hypoparathyroidism. Nature Reviews Disease Primers, 2017, 3, 17055.	18.1	142
7	Calcium-sensing receptor (CaSR) mutations and disorders of calcium, electrolyte and water metabolism. Best Practice and Research in Clinical Endocrinology and Metabolism, 2013, 27, 359-371.	2.2	118
8	Medial Arterial Calcification. Journal of the American College of Cardiology, 2021, 78, 1145-1165.	1.2	106
9	Hypercalcemic Disorders in Children. Journal of Bone and Mineral Research, 2017, 32, 2157-2170.	3.1	82
10	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.	1.9	73
11	Familial isolated primary hyperparathyroidism caused by mutations of the MEN1 gene. Nature Clinical Practice Endocrinology and Metabolism, 2008, 4, 53-58.	2.9	72
12	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
13	Unmet therapeutic, educational and scientific needs in parathyroid disorders: Consensus Statement from the first European Society of Endocrinology Workshop (PARAT). European Journal of Endocrinology, 2019, 181, P1-P19.	1.9	61
14	International Union of Basic and Clinical Pharmacology. CVIII. Calcium-Sensing Receptor Nomenclature, Pharmacology, and Function. Pharmacological Reviews, 2020, 72, 558-604.	7.1	59
15	Asymmetric activation of the calcium-sensing receptor homodimer. Nature, 2021, 595, 455-459.	13.7	59
16	The Calcilytic Agent NPS 2143 Rectifies Hypocalcemia in a Mouse Model With an Activating Calcium-Sensing Receptor (CaSR) Mutation: Relevance to Autosomal Dominant Hypocalcemia Type 1 (ADH1). Endocrinology, 2015, 156, 3114-3121.	1.4	55
17	A homozygous inactivating calciumâ€sensing receptor mutation, Pro339Thr, is associated with isolated primary hyperparathyroidism: correlation between location of mutations and severity of hypercalcaemia. Clinical Endocrinology, 2010, 73, 715-722.	1.2	53
18	Functional characterization of calcium sensing receptor polymorphisms and absence of association with indices of calcium homeostasis and bone mineral density. Clinical Endocrinology, 2006, 65, 598-605.	1.2	47

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19	Familial Hypocalciuric Hypercalcemia Type 1 and Autosomal-Dominant Hypocalcemia Type 1: Prevalence in a Large Healthcare Population. American Journal of Human Genetics, 2020, 106, 734-747.	2.6	45
20	Investigating hypocalcaemia. BMJ, The, 2013, 346, f2213-f2213.	3.0	44
21	A G-protein Subunit-Î±11 Loss-of-Function Mutation, Thr54Met, Causes Familial Hypocalciuric Hypercalcemia Type 2 (FHH2). Journal of Bone and Mineral Research, 2016, 31, 1200-1206.	3.1	40
22	Cinacalcet for Symptomatic Hypercalcemia Caused by AP2S1 Mutations. New England Journal of Medicine, 2016, 374, 1396-1398.	13.9	38
23	Identification of a G-Protein Subunit-Î±11 Gain-of-Function Mutation, Val340Met, in a Family With Autosomal Dominant Hypocalcemia Type 2 (ADH2). Journal of Bone and Mineral Research, 2016, 31, 1207-1214.	3.1	36
24	Cinacalcet Rectifies Hypercalcemia in a Patient With Familial Hypocalciuric Hypercalcemia Type 2 (FHH2) Caused by a Germline Loss-of-Function GÎ±11 Mutation. Journal of Bone and Mineral Research, 2018, 33, 32-41.	3.1	36
25	Autosomal Dominant Hypercalciuria in a Mouse Model Due to a Mutation of the Epithelial Calcium Channel, TRPV5. PLoS ONE, 2013, 8, e55412.	1.1	35
26	Identification of a Second Kindred with Familial Hypocalciuric Hypercalcemia Type 3 (FHH3) Narrows Localization to a <3.5 Megabase Pair Region on Chromosome 19q13.3. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 1947-1954.	1.8	34
27	A calcium-sensing receptor mutation causing hypocalcemia disrupts a transmembrane salt bridge to activate Î²-arrestinâ€“biased signaling. Science Signaling, 2018, 11, .	1.6	32
28	Oncogenic hypophosphataemic osteomalacia: biomarker roles of fibroblast growth factor 23, 1,25-dihydroxyvitamin D3 and lymphatic vessel endothelial hyaluronan receptor 1. European Journal of Endocrinology, 2008, 158, 265-271.	1.9	31
29	Allosteric Modulation of the Calcium-sensing Receptor Rectifies Signaling Abnormalities Associated with G-protein Î±-11 Mutations Causing Hypercalcemic and Hypocalcemic Disorders. Journal of Biological Chemistry, 2016, 291, 10876-10885.	1.6	31
30	Mutant Mice With Calcium-Sensing Receptor Activation Have Hyperglycemia That Is Rectified by Calcilytic Therapy. Endocrinology, 2017, 158, 2486-2502.	1.4	31
31	Calcimimetic and calcilytic therapies for inherited disorders of the calciumâ€sensing receptor signalling pathway. British Journal of Pharmacology, 2018, 175, 4083-4094.	2.7	29
32	An N-Ethyl-N-Nitrosourea Induced Corticotropin-Releasing Hormone Promoter Mutation Provides a Mouse Model for Endogenous Glucocorticoid Excess. Endocrinology, 2014, 155, 908-922.	1.4	28
33	GÎ±11 mutation in mice causes hypocalcemia rectifiable by calcilytic therapy. JCI Insight, 2017, 2, e91103.	2.3	28
34	Calcium-sensing receptor residues with loss- and gain-of-function mutations are located in regions of conformational change and cause signalling bias. Human Molecular Genetics, 2018, 27, 3720-3733.	1.4	23
35	Genetic approaches to metabolic bone diseases. British Journal of Clinical Pharmacology, 2019, 85, 1147-1160.	1.1	21
36	Mutational Analysis of the Adaptor Protein 2 Sigma Subunit (AP2S1) Gene: Search for Autosomal Dominant Hypocalcemia Type 3 (ADH3). Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1300-E1305.	1.8	19

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37	Role of Ca2+ and L-Phe in Regulating Functional Cooperativity of Disease-Associated â€œToggleâ€• Calcium-Sensing Receptor Mutations. PLoS ONE, 2014, 9, e113622.	1.1	18
38	Cinacalcet corrects hypercalcemia in mice with an inactivating GÎ±11 mutation. JCI Insight, 2017, 2, .	2.3	17
39	N-ethyl-N-Nitrosourea (ENU) Induced Mutations within the Klotho Gene Lead to Ectopic Calcification and Reduced Lifespan in Mouse Models. PLoS ONE, 2015, 10, e0122650.	1.1	16
40	Association Studies of Calcium-Sensing Receptor (CaSR) Polymorphisms with Serum Concentrations of Glucose and Phosphate, and Vascular Calcification in Renal Transplant Recipients. PLoS ONE, 2015, 10, e0119459.	1.1	15
41	Large-scale exome datasets reveal a new class of adaptor-related protein complex 2 sigma subunit (AP2Ïƒ) mutations, located at the interface with the AP2 alpha subunit, that impair calcium-sensing receptor signalling. Human Molecular Genetics, 2018, 27, 901-911.	1.4	15
42	Reference interval for albuminâ€adjusted calcium based on a large UK population. Clinical Endocrinology, 2021, 94, 34-39.	1.2	15
43	Vitamin D deficiency masking primary hyperparathyroidism. Annals of Clinical Biochemistry, 2004, 41, 405-407.	0.8	11
44	PTH Infusion for Seizures in Autosomal Dominant Hypocalcemia Type 1. New England Journal of Medicine, 2021, 385, 189-191.	13.9	11
45	Ap2s1 mutation causes hypercalcaemia in mice and impairs interaction between calcium-sensing receptor and adaptor protein-2. Human Molecular Genetics, 2021, 30, 880-892.	1.4	10
46	Comparison of human chromosome 19q13 and syntenic region on mouse chromosome 7 reveals absence, in man, of 11.6â€‰Mb containing four mouse calcium-sensing receptor-related sequences: relevance to familial benign hypocalciuric hypercalcaemia type 3. European Journal of Human Genetics, 2010, 18, 442-447.	1.4	8
47	Identification of a novel loss-of-function PHEX mutation, Ala720Ser, in a sporadic case of adult-onset hypophosphatemic osteomalacia. Bone, 2018, 106, 30-34.	1.4	8
48	Mice with a Brd4 Mutation Represent a New Model of Nephrocalcinosis. Journal of Bone and Mineral Research, 2019, 34, 1324-1335.	3.1	7
49	Neonatal Hypocalcemic Seizures in Offspring of a Mother With Familial Hypocalciuric Hypercalcemia Type 1 (FHH1). Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1393-1400.	1.8	7
50	Genetics of monogenic disorders of calcium and bone metabolism. Clinical Endocrinology, 2022, 97, 483-501.	1.2	7
51	Activating Mutations of the G-protein Subunit Î±â€Š11 Interdomain Interface Cause Autosomal Dominant Hypocalcemia Type 2. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 952-963.	1.8	6
52	Autosomal Dominant Hypocalcemia Type 1 (ADH1) Associated With Myoclonus and Intracerebral Calcifications. Journal of the Endocrine Society, 2022, 6, bvac042.	0.1	5
53	Calcilytic NPSP795 Increases Plasma Calcium and PTH in an Autosomal Dominant Hypocalcemia Type 1 Mouse Model. JBMR Plus, 2020, 4, e10402.	1.3	3
54	Genetics of Skeletal Disorders. Handbook of Experimental Pharmacology, 2020, 262, 325-351.	0.9	3

#	Article	IF	Citations
55	Case report: a 10-year-old girl with primary hypoparathyroidism and systemic lupus erythematosus. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1231-1235.	0.4	2
56	Calcium-sensing receptor (version 2019.4) in the IUPHAR/BPS Guide to Pharmacology Database. IUPHAR/BPS Guide To Pharmacology CITE, 2019, 2019, .	0.2	2
57	Spectrum of germline AIRE mutations causing APS-1 and familial hypoparathyroidism. European Journal of Endocrinology, 2022, , .	1.9	1
58	Hypoparathyroidism. , 2018, , 617-636.		0
59	Genetic regulation of parathyroid gland development. , 2020, , 1355-1377.		0