Rajesh V Thakker

List of Publications by Year
in descending order

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		8749	9090
271	23,076	75	144
papers	citations	h-index	g-index

281	281	281	15904
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	CONSENSUS: Guidelines for Diagnosis and Therapy of MEN Type 1 and Type 2. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5658-5671.	1.8	1,782
2	Gastroenteropancreatic neuroendocrine tumours. Lancet Oncology, The, 2008, 9, 61-72.	5.1	1,474
3	Clinical Practice Guidelines for Multiple Endocrine Neoplasia Type 1 (MEN1). Journal of Clinical Endocrinology and Metabolism, 2012, 97, 2990-3011.	1.8	1,127
4	Guidelines for the management of thyroid cancer. Clinical Endocrinology, 2014, 81, 1-122.	1.2	961
5	A common molecular basis for three inherited kidney stone diseases. Nature, 1996, 379, 445-449.	13.7	694
6	Multiple endocrine neoplasia type 1 (MEN1): analysis of 1336 mutations reported in the first decade following identification of the gene. Human Mutation, 2008, 29, 22-32.	1.1	591
7	CONSENSUS: Guidelines for Diagnosis and Therapy of MEN Type 1 and Type 2. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5658-5671.	1.8	574
8	A Familial Syndrome of Hypocalcemia with Hypercalciuria Due to Mutations in the Calcium-Sensing Receptor. New England Journal of Medicine, 1996, 335, 1115-1122.	13.9	565
9	GATA3 haplo-insufficiency causes human HDR syndrome. Nature, 2000, 406, 419-422.	13.7	516
10	Mutations Affecting G-Protein Subunit Î±₁₁in Hypercalcemia and Hypocalcemia. New England Journal of Medicine, 2013, 368, 2476-2486.	13.9	340
11	Association of Parathyroid Tumors in Multiple Endocrine Neoplasia Type 1 with Loss of Alleles on Chromosome 11. New England Journal of Medicine, 1989, 321, 218-224.	13.9	336
12	Multiple endocrine neoplasia type 1 (MEN1) and type 4 (MEN4). Molecular and Cellular Endocrinology, 2014, 386, 2-15.	1.6	334
13	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nature Genetics, 2015, 47, 717-726.	9.4	310
14	Management of Hypoparathyroidism: Summary Statement and Guidelines. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2273-2283.	1.8	303
15	Diagnosis of Asymptomatic Primary Hyperparathyroidism: Proceedings of the Fourth International Workshop. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 3570-3579.	1.8	296
16	Mice lacking renal chloride channel, CLC-5, are a model for Dent's disease, a nephrolithiasis disorder associated with defective receptor-mediated endocytosis. Human Molecular Genetics, 2000, 9, 2937-2945.	1.4	273
17	Presentation of Hypoparathyroidism: Etiologies and Clinical Features. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2300-2312.	1.8	246
18	Mutations in AP2S1 cause familial hypocalciuric hypercalcemia type 3. Nature Genetics, 2013, 45, 93-97.	9.4	242

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19	Genome-Wide Association Study Using Extreme Truncate Selection Identifies Novel Genes Affecting Bone Mineral Density and Fracture Risk. PLoS Genetics, 2011, 7, e1001372.	1.5	233
20	Epidemiology and Diagnosis of Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2284-2299.	1.8	230
21	The calcium-sensing receptor in physiology and in calcitropic and noncalcitropic diseases. Nature Reviews Endocrinology, 2019, 15, 33-51.	4.3	226
22	Genetic Disorders of Renal Electrolyte Transport. New England Journal of Medicine, 1999, 340, 1177-1187.	13.9	224
23	A donor splice site mutation in the parathyroid hormone gene is associated with autosomal recessive hypoparathyroidism. Nature Genetics, 1992, 1, 149-152.	9.4	211
24	Glomerular protein sieving and implications for renal failure in Fanconi syndrome. Kidney International, 2001, 60, 1885-1892.	2.6	207
25	Multiple endocrine neoplasia type 1 (MEN1). Best Practice and Research in Clinical Endocrinology and Metabolism, 2010, 24, 355-370.	2.2	206
26	Dent's disease. Orphanet Journal of Rare Diseases, 2010, 5, 28.	1.2	181
27	Cell division cycle protein 73 homolog (CDC73) mutations in the hyperparathyroidism-jaw tumor syndrome (HPT-JT) and parathyroid tumors. Human Mutation, 2010, 31, 295-307.	1.1	169
28	Autosomal Dominant Familial Hypoparathyroidism, Sensorineural Deafness, and Renal Dysplasia. New England Journal of Medicine, 1992, 327, 1069-1074.	13.9	161
29	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
30	Cloning and Characterization of CLCN5, the Human Kidney Chloride Channel Gene Implicated in Dent Disease (an X-Linked Hereditary Nephrolithiasis). Genomics, 1995, 29, 598-606.	1.3	148
31	Characterisation of renal chloride channel, CLCN5, mutations in hypercalciuric nephrolithiasis (kidney stones) disorders. Human Molecular Genetics, 1997, 6, 1233-1239.	1.4	148
32	Characterization of GATA3 Mutations in the Hypoparathyroidism, Deafness, and Renal Dysplasia (HDR) Syndrome. Journal of Biological Chemistry, 2004, 279, 22624-22634.	1.6	145
33	Heterogeneous Genetic Background of the Association of Pheochromocytoma/Paraganglioma and Pituitary Adenoma: Results From a Large Patient Cohort. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E531-E541.	1.8	145
34	Landscape of Familial Isolated and Young-Onset Pituitary Adenomas: Prospective Diagnosis inAIPMutation Carriers. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1242-E1254.	1.8	144
35	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
36	Hypoparathyroidism. Nature Reviews Disease Primers, 2017, 3, 17055.	18.1	142

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37	An interstitial deletion-insertion involving chromosomes 2p25.3 and Xq27.1, near SOX3, causes X-linked recessive hypoparathyroidism. Journal of Clinical Investigation, 2005, 115, 2822-2831.	3.9	135
38	Multiple Endocrine Neoplasiaâ€"Syndromes of the Twentieth Century. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 2617-2620.	1.8	131
39	Localization of Familial Benign Hypercalcemia, Oklahoma Variant (FBHOk), to Chromosome 19q13. American Journal of Human Genetics, 1999, 64, 189-195.	2.6	129
40	Functional characterization of GATA3 mutations causing the hypoparathyroidism-deafness-renal (HDR) dysplasia syndrome: insight into mechanisms of DNA binding by the GATA3 transcription factor. Human Molecular Genetics, 2006, 16, 265-275.	1.4	129
41	Activating calcium-sensing receptor mutation in the mouse is associated with cataracts and ectopic calcification. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 13566-13571.	3.3	126
42	Whole-Exome Sequencing Studies of Nonhereditary (Sporadic) Parathyroid Adenomas. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1995-E2005.	1.8	121
43	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
44	Gata3-deficient mice develop parathyroid abnormalities due to dysregulation of the parathyroid-specific transcription factor Gcm2. Journal of Clinical Investigation, 2010, 120, 2144-2155.	3.9	108
45	Mutant Prolactin Receptor and Familial Hyperprolactinemia. New England Journal of Medicine, 2013, 369, 2012-2020.	13.9	106
46	Medial Arterial Calcification. Journal of the American College of Cardiology, 2021, 78, 1145-1165.	1.2	106
47	Mutational Analysis of PHEX Gene in X-Linked Hypophosphatemia1. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 3615-3623.	1.8	104
48	Genetic contribution to renal function and electrolyte balance: a twin study. Clinical Science, 2002, 103, 259-265.	1.8	104
49	GNAS Mutations in Pseudohypoparathyroidism Type 1a and Related Disorders. Human Mutation, 2015, 36, 11-19.	1.1	101
50	Urinary Megalin Deficiency Implicates Abnormal Tubular Endocytic Function in Fanconi Syndrome. Journal of the American Society of Nephrology: JASN, 2002, 13, 125-133.	3.0	101
51	Dent's disease, a renal Fanconi syndrome with nephrocalcinosis and kidney stones, is associated with a microdeletion involving DXS255 and maps to Xp11.22. Human Molecular Genetics, 1993, 2, 2129-2134.	1.4	100
52	Anatomic and Functional Imaging of Metastatic Carcinoid Tumors. Radiographics, 2007, 27, 455-477.	1.4	100
53	Tubular proteinuria defined by a study of Dent's (CLCN5 mutation) and other tubular diseases. Kidney International, 2000, 57, 240-249.	2.6	99
54	Parafibromin, a Component of the Human PAF Complex, Regulates Growth Factors and Is Required for Embryonic Development and Survival in Adult Mice. Molecular and Cellular Biology, 2008, 28, 2930-2940.	1.1	97

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55	Genetic causes of hypercalciuric nephrolithiasis. Pediatric Nephrology, 2009, 24, 2321-2332.	0.9	97
56	Uromodulin mutations causing familial juvenile hyperuricaemic nephropathy lead to protein maturation defects and retention in the endoplasmic reticulum. Human Molecular Genetics, 2009, 18, 2963-2974.	1.4	94
57	Pathogenesis of Dent's disease and related syndromes of X-linked nephrolithiasis. Kidney International, 2000, 57, 787-793.	2.6	92
58	Menin Interacts Directly with the Homeobox-Containing Protein Pem. Biochemical and Biophysical Research Communications, 2001, 286, 426-431.	1.0	92
59	Neonatal severe hyperparathyroidism: genotype/phenotype correlation and the use of pamidronate as rescue therapy. European Journal of Pediatrics, 2004, 163, 589-594.	1.3	92
60	MMP13 mutation causes spondyloepimetaphyseal dysplasia, Missouri type (SEMDMO). Journal of Clinical Investigation, 2005, 115, 2832-2842.	3.9	89
61	Asymptomatic Children with Multiple Endocrine Neoplasia Type 1 Mutations May Harbor Nonfunctioning Pancreatic Neuroendocrine Tumors. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 3640-3646.	1.8	88
62	Multiple endocrine neoplasia type 1 knockout mice develop parathyroid, pancreatic, pituitary and adrenal tumours with hypercalcaemia, hypophosphataemia and hypercorticosteronaemia. Endocrine-Related Cancer, 2009, 16, 1313-1327.	1.6	88
63	Whole-Exome Sequencing Studies of Parathyroid Carcinomas Reveal NovelPRUNE2Mutations, Distinctive Mutational Spectra Related to APOBEC-Catalyzed DNA Mutagenesis and Mutational Enrichment in Kinases Associated With Cell Migration and Invasion. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E360-E364.	1.8	86
64	Whole-Exome Sequencing Studies of Nonfunctioning Pituitary Adenomas. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E796-E800.	1.8	85
65	Multiple Endocrine Neoplasia Type 1: Latest Insights. Endocrine Reviews, 2021, 42, 133-170.	8.9	85
66	Genomic profiling reveals mutational landscape in parathyroid carcinomas. JCI Insight, 2017, 2, e92061.	2.3	84
67	Clinical Features of Multiple Endocrine Neoplasia Type 4: Novel Pathogenic Variant and Review of Published Cases. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3637-3646.	1.8	83
68	Mutations of CLCN5 in Japanese children with idiopathic low molecular weight proteinuria, hypercalciuria and nephrocalcinosis. Kidney International, 1997, 52, 911-916.	2.6	82
69	Molecular genetics of syndromic and non-syndromic forms of parathyroid carcinoma. Human Mutation, 2017, 38, 1621-1648.	1.1	82
70	Hypercalcemic Disorders in Children. Journal of Bone and Mineral Research, 2017, 32, 2157-2170.	3.1	82
71	Challenges and controversies in management of pancreatic neuroendocrine tumours in patients with MEN1. Lancet Diabetes and Endocrinology,the, 2015, 3, 895-905.	5.5	81
72	Standards of care for hypoparathyroidism in adults: a Canadian and International Consensus. European Journal of Endocrinology, 2019, 180, P1-P22.	1.9	81

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73	Genetics of kidney stone disease. Nature Reviews Urology, 2020, 17, 407-421.	1.9	81
74	MULTIPLE ENDOCRINE NEOPLASIA TYPE 1. Endocrinology and Metabolism Clinics of North America, 2000, 29, 541-567.	1.2	80
75	Somatic Mutations in MEN Type 1 Tumors, Consistent with the Knudson â€œTwo-Hitâ€•Hypothesis. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 4371-4374.	1.8	79
76	<i>OCRL1</i> Mutations in Dent 2 Patients Suggest a Mechanism for Phenotypic Variability. Nephron Physiology, 2009, 112, p27-p36.	1.5	79
77	Diagnostic challenges due to phenocopies: lessons from Multiple Endocrine Neoplasia type1 (MEN1). Human Mutation, 2010, 31, E1089-E1101.	1.1	78
78	Constitutional deletion of chromosome 20q in two patients affected with albright hereditary osteodystrophy. American Journal of Medical Genetics Part A, 2002, 113, 167-172.	2.4	77
79	Definition of the MinimalMEN1Candidate Area Based on a 5-Mb Integrated Map of Proximal 11q13. Genomics, 1996, 37, 354-365.	1.3	76
80	Altered polarity and expression of H+-ATPase without ultrastructural changes in kidneys of Dent's disease patients. Kidney International, 2003, 63, 1285-1295.	2.6	76
81	The ClC-5 Knockout Mouse Model of Dent's Disease Has Renal Hypercalciuria and Increased Bone Turnover. Journal of Bone and Mineral Research, 2003, 18, 615-623.	3.1	75
82	Familial isolated primary hyperparathyroidism caused by mutations of the MEN1 gene. Nature Clinical Practice Endocrinology and Metabolism, 2008, 4, 53-58.	2.9	72
83	Functional characterization of renal chloride channel, CLCN5, mutations associated with Dentâ€™sJapan disease. Kidney International, 1998, 54, 1850-1856.	2.6	71
84	Role of Multiple Endocrine Neoplasia Type 1 Mutational Analysis in Clinical Practice. Endocrine Practice, 2011, 17, 8-17.	1.1	71
85	Receptor-mediated endocytosis and endosomal acidification is impaired in proximal tubule epithelial cells of Dent disease patients. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 7014-7019.	3.3	71
86	Membrane Targeting and Secretion of Mutant Uromodulin in Familial Juvenile Hyperuricemic Nephropathy. Journal of the American Society of Nephrology: JASN, 2007, 18, 264-273.	3.0	70
87	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
88	Genetic variants of calcium and vitamin D metabolism in kidney stone disease. Nature Communications, 2019, 10, 5175.	5.8	69
89	Multiple Endocrine Neoplasia: Spectrum of Radiologic Appearances and Discussion of a Multitechnique Imaging Approach. Radiographics, 2006, 26, 433-451.	1.4	67
90	AP2Ïƒ Mutations Impair Calcium-Sensing Receptor Trafficking and Signaling, and Show an Endosomal Pathway to Spatially Direct G-Protein Selectivity. Cell Reports, 2018, 22, 1054-1066.	2.9	66

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91	Isolated hypercalciuria with mutation in CLCN5: Relevance to idiopathic hypercalciuria. Kidney International, 2000, 57, 232-239.	2.6	65
92	Hyperparathyroidism-Jaw Tumor Syndrome in Roma Families from Portugal Is Due to a Founder Mutation of the HRPT2 Gene. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 1747-1752.	1.8	65
93	Multiple Endocrine Neoplasia Type 1 and the Pancreas: Diagnosis and Treatment of Functioning and Non-Functioning Pancreatic and Duodenal Neuroendocrine Neoplasia within the MEN1 Syndrome â€“ An International Consensus Statement. Neuroendocrinology, 2021, 111, 609-630.	1.2	63
94	Expression and Cloning of the Human X-Linked Hypophosphatemia Gene cDNA. Biochemical and Biophysical Research Communications, 1997, 231, 635-639.	1.0	62
95	Linkage studies in a kindred from Oklahoma, with familial benign (hypocalciuric) hypercalcaemia (FBH) and developmental elevations in serum parathyroid hormone levels, indicate a third locus for FBH. Human Genetics, 1995, 96, 183-187.	1.8	61
96	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
97	International Union of Basic and Clinical Pharmacology. CVIII. Calcium-Sensing Receptor Nomenclature, Pharmacology, and Function. Pharmacological Reviews, 2020, 72, 558-604.	7.1	59
98	Asymmetric activation of the calcium-sensing receptor homodimer. Nature, 2021, 595, 455-459.	13.7	59
99	Functional Analysis of a Novel GATA3 Mutation in a Family with the Hypoparathyroidism, Deafness, and Renal Dysplasia Syndrome. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 2445-2450.	1.8	58
100	Significant deterioration in nanomechanical quality occurs through incomplete extrafibrillar mineralization in rachitic bone: Evidence from in-situ synchrotron X-ray scattering and backscattered electron imaging. Journal of Bone and Mineral Research, 2012, 27, 876-890.	3.1	58
101	Association of familial duane anomaly and urogenital abnormalities with a bisatellited marker derived from chromosome 22. American Journal of Medical Genetics Part A, 1993, 47, 925-930.	2.4	57
102	Establishing normal plasma and 24-hour urinary biochemistry ranges in C3H, BALB/c and C57BL/6J mice following acclimatization in metabolic cages. Laboratory Animals, 2010, 44, 218-225.	0.5	57
103	CLC-5 and KIF3B interact to facilitate CLC-5 plasma membrane expression, endocytosis, and microtubular transport: relevance to pathophysiology of Dent's disease. American Journal of Physiology - Renal Physiology, 2010, 298, F365-F380.	1.3	56
104	Genetics of Hypercalciuric Nephrolithiasis. Annals of the New York Academy of Sciences, 2007, 1116, 461-484.	1.8	55
105	A MissenseGlial Cells Missing Homolog B(GCMB) Mutation, Asn502His, Causes Autosomal Dominant Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3512-3516.	1.8	55
106	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
107	Clinical and genetic studies of CLCN5 mutations in Japanese families with Dent's disease. Kidney International, 2000, 58, 520-527.	2.6	54
108	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

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109	Frequent Occurrence of an Intron 4 Mutation in Multiple Endocrine Neoplasia Type 1. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 2688-2693.	1.8	51
110	Association between Genotype and Phenotype in Uromodulin-Associated Kidney Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2013, 8, 1349-1357.	2.2	51
111	Hormonal regulation of biomineralization. Nature Reviews Endocrinology, 2021, 17, 261-275.	4.3	50
112	Modeling study of human renal chloride channel (hCLC-5) mutations suggests a structural-functional relationship. Kidney International, 2003, 63, 1426-1432.	2.6	49
113	Identification and characterization of novel parathyroid-specific transcription factor Glial Cells Missing Homolog B (GCMB) mutations in eight families with autosomal recessive hypoparathyroidism. Human Molecular Genetics, 2010, 19, 2028-2038.	1.4	48
114	X-Linked Hypophosphatemia Attributable to Pseudoexons of the PHEX Gene. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 3840-3844.	1.8	47
115	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
116	Characterization of Dent's disease mutations of CLC-5 reveals a correlation between functional and cell biological consequences and protein structure. American Journal of Physiology - Renal Physiology, 2009, 296, F390-F397.	1.3	47
117	Transcription factors in parathyroid development: lessons from hypoparathyroid disorders. Annals of the New York Academy of Sciences, 2011, 1237, 24-38.	1.8	47
118	CDC73 Intragenic Deletion in Familial Primary Hyperparathyroidism Associated With Parathyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 3044-3048.	1.8	46
119	Current and emerging therapies for PNETs in patients with or without MEN1. Nature Reviews Endocrinology, 2018, 14, 216-227.	4.3	46
120	The role of biomineralization in disorders of skeletal development and tooth formation. Nature Reviews Endocrinology, 2021, 17, 336-349.	4.3	46
121	Characterization of Renal Chloride Channel (CLCN5) Mutations in Dent's Disease. Journal of the American Society of Nephrology: JASN, 2000, 11, 1460-1468.	3.0	46
122	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
123	Investigating hypocalcaemia. BMJ, The, 2013, 346, f2213-f2213.	3.0	44
124	Quantitative Trait Loci for Hypercalciuria in a Rat Model of Kidney Stone Disease. Journal of the American Society of Nephrology: JASN, 2003, 14, 1844-1850.	3.0	42
125	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
126	Genetic background influences embryonic lethality and the occurrence of neural tube defects in Men1 null mice: relevance to genetic modifiers. Journal of Endocrinology, 2009, 203, 133-142.	1.2	38

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127	Cinacalcet for Symptomatic Hypercalcemia Caused by AP2S1 Mutations. New England Journal of Medicine, 2016, 374, 1396-1398.	13.9	38
128	Somatic mutations of GNA11 and GNAQ in CTNNB1-mutant aldosterone-producing adenomas presenting in puberty, pregnancy or menopause. Nature Genetics, 2021, 53, 1360-1372.	9.4	37
129	Clinical features of X-linked nephrolithiasis in childhood. Pediatric Nephrology, 1998, 12, 625-629.	0.9	36
130	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
131	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
132	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
133	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
134	MEN1 Gene Replacement Therapy Reduces Proliferation Rates in a Mouse Model of Pituitary Adenomas. Cancer Research, 2012, 72, 5060-5068.	0.4	34
135	Mouse model for inherited renal fibrosis associated with endoplasmic reticulum stress. DMM Disease Models and Mechanisms, 2017, 10, 773-786.	1.2	34
136	A Missense GATA3 Mutation, Thr272Ile, Causes the Hypoparathyroidism, Deafness, and Renal Dysplasia Syndrome. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 3897-3904.	1.8	33
137	Renal Chloride Channel, CLCN5, Mutations in Dent's Disease. Journal of Bone and Mineral Research, 1999, 14, 1536-1542.	3.1	32
138	miR-135b- and miR-146b-dependent silencing of calcium-sensing receptor expression in colorectal tumors. International Journal of Cancer, 2016, 138, 137-145.	2.3	32
139	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
140	Construction of a 1.2-Mb Sequence-Ready Contig of Chromosome 11q13 Encompassing the Multiple Endocrine Neoplasia Type 1 (MEN1) Gene. Genomics, 1997, 44, 94-100.	1.3	31
141	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
142	A mouse model for spondyloepiphyseal dysplasia congenita with secondary osteoarthritis due to a Col2a1 mutation. Journal of Bone and Mineral Research, 2012, 27, 413-428.	3.1	31
143	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
144	Mutant Mice With Calcium-Sensing Receptor Activation Have Hyperglycemia That Is Rectified by Calcilytic Therapy. Endocrinology, 2017, 158, 2486-2502.	1.4	31

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145	A Novel Role for GATA3 in Mesangial Cells in Glomerular Development and Injury. Journal of the American Society of Nephrology: JASN, 2019, 30, 1641-1658.	3.0	31
146	Mutational Analysis of CLC-5, Cofilin and CLC-4 in Patients with Dentâ€™s Disease. Nephron Physiology, 2009, 112, p53-p62.	1.5	30
147	Mouse models for inherited endocrine and metabolic disorders. Journal of Endocrinology, 2011, 211, 211-230.	1.2	30
148	Confusing genes: a patient with MEN2A and Cushing's disease. Clinical Endocrinology, 2013, 78, 966-968.	1.2	30
149	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
150	Chloride channels cough up. Nature Genetics, 1997, 17, 125-127.	9.4	28
151	GATA3 Mutations Found in Breast Cancers May Be Associated with Aberrant Nuclear Localization, Reduced Transactivation and Cell Invasiveness. Hormones and Cancer, 2013, 4, 123-139.	4.9	28
152	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
153	GÎ±11 mutation in mice causes hypocalcemia rectifiable by calcilytic therapy. JCI Insight, 2017, 2, e91103.	2.3	28
154	Comparative ontogeny, processing, and segmental distribution of the renal chloride channel, ClC-5. Kidney International, 2004, 65, 198-208.	2.6	27
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