Rajesh V Thakker

List of Publications by Citations

262 papers	17,766 citations	67 h-index	128 g-index
279 ext. papers	20,126 ext. citations	9.5 avg, IF	6.59 L-index

#	Paper	IF	Citations
262	Guidelines for diagnosis and therapy of MEN type 1 and type 2. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 5658-71	5.6	1466
261	Gastroenteropancreatic neuroendocrine tumours. *Lancet Oncology, The*, **2008**, 9, 61-72	21.7	1246
260	Clinical practice guidelines for multiple endocrine neoplasia type 1 (MEN1). *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, 2990-3011	5.6	830
259	Guidelines for the management of thyroid cancer. *Clinical Endocrinology*, **2014**, 81 Suppl 1, 1-122	3.4	692
258	A common molecular basis for three inherited kidney stone diseases. *Nature*, **1996**, 379, 445-9	50.4	614
257	A familial syndrome of hypocalcemia with hypercalciuria due to mutations in the calcium-sensing receptor. *New England Journal of Medicine*, **1996**, 335, 1115-22	59.2	496
256	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	4.7	494
255	GATA3 haplo-insufficiency causes human HDR syndrome. *Nature*, **2000**, 406, 419-22	50.4	452
254	Mutations affecting G-protein subunit α11 in hypercalcemia and hypocalcemia. *New England Journal of Medicine*, **2013**, 368, 2476-2486	59.2	282
253	Association of parathyroid tumors in multiple endocrine neoplasia type 1 with loss of alleles on chromosome 11. *New England Journal of Medicine*, **1989**, 321, 218-24	59.2	280
252	Multiple endocrine neoplasia type 1 (MEN1) and type 4 (MEN4). *Molecular and Cellular Endocrinology*, **2014**, 386, 2-15	4.4	261
251	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, 2937-45	5.6	248
250	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. *Nature Genetics*, **2015**, 47, 717-726	36.3	244
249	Diagnosis of asymptomatic primary hyperparathyroidism: proceedings of the Fourth International Workshop. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, 3570-9	5.6	233
248	Management of Hypoparathyroidism: Summary Statement and Guidelines. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 2273-83	5.6	207
247	Genome-wide association study using extreme truncate selection identifies novel genes affecting bone mineral density and fracture risk. *PLoS Genetics*, **2011**, 7, e1001372	6	199
246	Mutations in AP2S1 cause familial hypocalciuric hypercalcemia type 3. *Nature Genetics*, **2013**, 45, 93-7	36.3	196

245	A donor splice site mutation in the parathyroid hormone gene is associated with autosomal recessive hypoparathyroidism. *Nature Genetics*, **1992**, 1, 149-52	36.3	196
244	Genetic disorders of renal electrolyte transport. *New England Journal of Medicine*, **1999**, 340, 1177-87	59.2	183
243	Presentation of Hypoparathyroidism: Etiologies and Clinical Features. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 2300-12	5.6	175
242	Glomerular protein sieving and implications for renal failure in Fanconi syndrome. *Kidney International*, **2001**, 60, 1885-92	9.9	173
241	Multiple endocrine neoplasia type 1 (MEN1). *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2010**, 24, 355-70	6.5	161
240	Epidemiology and Diagnosis of Hypoparathyroidism. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 2284-99	5.6	148
239	Brief report: autosomal dominant familial hypoparathyroidism, sensorineural deafness, and renal dysplasia. *New England Journal of Medicine*, **1992**, 327, 1069-74	59.2	141
238	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	4.7	138
237	Dent's disease. *Orphanet Journal of Rare Diseases*, **2010**, 5, 28	4.2	133
236	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, 2768-78	5.6	133
235	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	4.3	131
234	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, E531-41	5.6	127
233	Characterisation of renal chloride channel, CLCN5, mutations in hypercalciuric nephrolithiasis (kidney stones) disorders. *Human Molecular Genetics*, **1997**, 6, 1233-9	5.6	125
232	Characterization of GATA3 mutations in the hypoparathyroidism, deafness, and renal dysplasia (HDR) syndrome. *Journal of Biological Chemistry*, **2004**, 279, 22624-34	5.4	122
231	Localization of familial benign hypercalcemia, Oklahoma variant (FBHOk), to chromosome 19q13. *American Journal of Human Genetics*, **1999**, 64, 189-95	11	119
230	Disorders of the calcium-sensing receptor and partner proteins: insights into the molecular basis of calcium homeostasis. *Journal of Molecular Endocrinology*, **2016**, 57, R127-42	4.5	116
229	Landscape of Familial Isolated and Young-Onset Pituitary Adenomas: Prospective Diagnosis in AIP Mutation Carriers. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E1242-54	5.6	115
228	The calcium-sensing receptor in physiology and in calcitropic and noncalcitropic diseases. *Nature Reviews Endocrinology*, **2018**, 15, 33-51	15.2	113

227	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*, **2007**, 16, 265-75	5.6	111
226	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, 13566-71	11.5	111
225	An interstitial deletion-insertion involving chromosomes 2p25.3 and Xq27.1, near SOX3, causes X-linked recessive hypoparathyroidism. *Journal of Clinical Investigation*, **2005**, 115, 2822-31	15.9	110
224	Multiple endocrine neoplasia--syndromes of the twentieth century. *Journal of Clinical Endocrinology and Metabolism*, **1998**, 83, 2617-20	5.6	109
223	Whole-exome sequencing studies of nonhereditary (sporadic) parathyroid adenomas. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E1995-2005	5.6	101
222	Hypoparathyroidism. *Nature Reviews Disease Primers*, **2017**, 3, 17055	51.1	100
221	Calcium-sensing receptor (CaSR) mutations and disorders of calcium, electrolyte and water metabolism. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2013**, 27, 359-71	6.5	98
220	Gata3-deficient mice develop parathyroid abnormalities due to dysregulation of the parathyroid-specific transcription factor Gcm2. *Journal of Clinical Investigation*, **2010**, 120, 2144-55	15.9	95
219	Mutational analysis of PHEX gene in X-linked hypophosphatemia. *Journal of Clinical Endocrinology and Metabolism*, **1998**, 83, 3615-23	5.6	94
218	Urinary megalin deficiency implicates abnormal tubular endocytic function in Fanconi syndrome. *Journal of the American Society of Nephrology: JASN*, **2002**, 13, 125-133	12.7	90
217	Menin interacts directly with the homeobox-containing protein Pem. *Biochemical and Biophysical Research Communications*, **2001**, 286, 426-31	3.4	88
216	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, 2129-34	5.6	88
215	Anatomic and functional imaging of metastatic carcinoid tumors. *Radiographics*, **2007**, 27, 455-77	5.4	87
214	Neonatal severe hyperparathyroidism: genotype/phenotype correlation and the use of pamidronate as rescue therapy. *European Journal of Pediatrics*, **2004**, 163, 589-94	4.1	87
213	Genetic contribution to renal function and electrolyte balance: a twin study. *Clinical Science*, **2002**, 103, 259-65	6.5	86
212	Tubular proteinuria defined by a study of Dent's (CLCN5 mutation) and other tubular diseases. *Kidney International*, **2000**, 57, 240-9	9.9	83
211	GNAS mutations in Pseudohypoparathyroidism type 1a and related disorders. *Human Mutation*, **2015**, 36, 11-9	4.7	82
210	Uromodulin mutations causing familial juvenile hyperuricaemic nephropathy lead to protein maturation defects and retention in the endoplasmic reticulum. *Human Molecular Genetics*, **2009**, 18, 2963-74	5.6	81

209	Mutant prolactin receptor and familial hyperprolactinemia. *New England Journal of Medicine*, **2013**, 369, 2012-2020	59.2	80
208	Genetic causes of hypercalciuric nephrolithiasis. *Pediatric Nephrology*, **2009**, 24, 2321-32	3.2	79
207	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, 2930-40	4.8	78
206	Pathogenesis of Dent's disease and related syndromes of X-linked nephrolithiasis. *Kidney International*, **2000**, 57, 787-93	9.9	77
205	MMP13 mutation causes spondyloepimetaphyseal dysplasia, Missouri type (SEMD(MO). *Journal of Clinical Investigation*, **2005**, 115, 2832-42	15.9	74
204	Asymptomatic children with multiple endocrine neoplasia type 1 mutations may harbor nonfunctioning pancreatic neuroendocrine tumors. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 3640-6	5.6	73
203	Definition of the MinimalMEN1Candidate Area Based on a 5-Mb Integrated Map of Proximal 11q13. *Genomics*, **1996**, 37, 354-365	4.3	72
202	Multiple endocrine neoplasia type 1 knockout mice develop parathyroid, pancreatic, pituitary and adrenal tumours with hypercalcaemia, hypophosphataemia and hypercorticosteronaemia. *Endocrine-Related Cancer*, **2009**, 16, 1313-27	5.7	71
201	Mutations of CLCN5 in Japanese children with idiopathic low molecular weight proteinuria, hypercalciuria and nephrocalcinosis. *Kidney International*, **1997**, 52, 911-6	9.9	70
200	Somatic mutations in MEN type 1 tumors, consistent with the Knudson "two-hit" hypothesis. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 4371-4	5.6	70
199	Whole-exome sequencing studies of nonfunctioning pituitary adenomas. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, E796-800	5.6	69
198	Whole-exome sequencing studies of parathyroid carcinomas reveal novel PRUNE2 mutations, 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, E360-4	5.6	68
197	Constitutional deletion of chromosome 20q in two patients affected with albright hereditary osteodystrophy. *American Journal of Medical Genetics Part A*, **2002**, 113, 167-72		68
196	Altered polarity and expression of H+-ATPase without ultrastructural changes in kidneys of Dent's disease patients. *Kidney International*, **2003**, 63, 1285-95	9.9	67
195	Challenges and controversies in management of pancreatic neuroendocrine tumours in patients with MEN1. *Lancet Diabetes and Endocrinology,the*, **2015**, 3, 895-905	18.1	65
194	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, 615-23	6.3	65
193	Functional characterization of renal chloride channel, CLCN5, mutations associated with Dent'sJapan disease. *Kidney International*, **1998**, 54, 1850-6	9.9	63
192	OCRL1 mutations in Dent 2 patients suggest a mechanism for phenotypic variability. *Nephron Physiology*, **2009**, 112, p27-36		62

191	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, 7014-9	11.5	61
190	Familial isolated primary hyperparathyroidism caused by mutations of the MEN1 gene. *Nature Clinical Practice Endocrinology and Metabolism*, **2008**, 4, 53-8		61
189	Isolated hypercalciuria with mutation in CLCN5: relevance to idiopathic hypercalciuria. *Kidney International*, **2000**, 57, 232-9	9.9	61
188	Membrane targeting and secretion of mutant uromodulin in familial juvenile hyperuricemic nephropathy. *Journal of the American Society of Nephrology: JASN*, **2007**, 18, 264-73	12.7	60
187	Multiple endocrine neoplasia type 1. *Endocrinology and Metabolism Clinics of North America*, **2000**, 29, 541-67	5.5	60
186	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, 1747-52	5.6	58
185	Expression and cloning of the human X-linked hypophosphatemia gene cDNA. *Biochemical and Biophysical Research Communications*, **1997**, 231, 635-9	3.4	57
184	Role of multiple endocrine neoplasia type 1 mutational analysis in clinical practice. *Endocrine Practice*, **2011**, 17 Suppl 3, 8-17	3.2	56
183	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, 183-7	6.3	56
182	Molecular genetics of syndromic and non-syndromic forms of parathyroid carcinoma. *Human Mutation*, **2017**, 38, 1621-1648	4.7	55
181	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, 2445-50	5.6	54
180	Adaptor protein-2 sigma subunit mutations causing familial hypocalciuric hypercalcaemia type 3 (FHH3) demonstrate genotype-phenotype correlations, codon bias and dominant-negative effects. *Human Molecular Genetics*, **2015**, 24, 5079-92	5.6	53
179	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, 876-90	6.3	53
178	Diagnostic challenges due to phenocopies: lessons from Multiple Endocrine Neoplasia type1 (MEN1). *Human Mutation*, **2010**, 31, E1089-101	4.7	53
177	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, 925-30		53
176	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, F365-80	4.3	52
175	Hypercalcemic Disorders in Children. *Journal of Bone and Mineral Research*, **2017**, 32, 2157-2170	6.3	51
174	Multiple endocrine neoplasia: spectrum of radiologic appearances and discussion of a multitechnique imaging approach. *Radiographics*, **2006**, 26, 433-51	5.4	51

173	A missense glial cells missing homolog B (GCMB) mutation, Asn502His, causes autosomal dominant hypoparathyroidism. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, 3512-6	5.6	50
172	Genomic profiling reveals mutational landscape in parathyroid carcinomas. *JCI Insight*, **2017**, 2, e92061	9.9	50
171	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, 3114-21	4.8	48
170	Frequent occurrence of an intron 4 mutation in multiple endocrine neoplasia type 1. *Journal of Clinical Endocrinology and Metabolism*, **2002**, 87, 2688-93	5.6	47
169	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	5.6	46
168	Genetics of hypercalciuric nephrolithiasis: renal stone disease. *Annals of the New York Academy of Sciences*, **2007**, 1116, 461-84	6.5	46
167	Modeling study of human renal chloride channel (hCLC-5) mutations suggests a structural-functional relationship. *Kidney International*, **2003**, 63, 1426-32	9.9	46
166	AP2 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	10.6	44
165	A homozygous inactivating calcium-sensing receptor mutation, Pro339Thr, is associated with isolated primary hyperparathyroidism: correlation between location of mutations and severity of hypercalcaemia. *Clinical Endocrinology*, **2010**, 73, 715-22	3.4	44
164	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, 218-25	2.6	44
163	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, 2028-38	5.6	44
162	Clinical and genetic studies of CLCN5 mutations in Japanese families with Dent's disease. *Kidney International*, **2000**, 58, 520-7	9.9	44
161	Standards of care for hypoparathyroidism in adults: a Canadian and International Consensus. *European Journal of Endocrinology*, **2019**, 180, P1-P22	6.5	44
160	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, F390-7	4.3	43
159	Transcription factors in parathyroid development: lessons from hypoparathyroid disorders. *Annals of the New York Academy of Sciences*, **2011**, 1237, 24-38	6.5	42
158	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	3.4	42
157	MANAGEMENT OF ENDOCRINE DISEASE: Unmet therapeutic, educational and scientific needs in parathyroid disorders. *European Journal of Endocrinology*, **2019**, 181, P1-P19	6.5	40
156	Characterization of renal chloride channel (CLCN5) mutations in Dent's disease. *Journal of the American Society of Nephrology: JASN*, **2000**, 11, 1460-1468	12.7	40

155	CDC73 intragenic deletion in familial primary hyperparathyroidism associated with parathyroid carcinoma. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, 3044-8	5.6	39
154	Association between genotype and phenotype in uromodulin-associated kidney disease. *Clinical Journal of the American Society of Nephrology: CJASN*, **2013**, 8, 1349-57	6.9	39
153	X-linked hypophosphatemia attributable to pseudoexons of the PHEX gene. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 3840-4	5.6	39
152	Quantitative trait loci for hypercalciuria in a rat model of kidney stone disease. *Journal of the American Society of Nephrology: JASN*, **2003**, 14, 1844-50	12.7	38
151	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, 133-42	4.7	36
150	Current and emerging therapies for PNETs in patients with or without MEN1. *Nature Reviews Endocrinology*, **2018**, 14, 216-227	15.2	34
149	Investigating hypocalcaemia. *BMJ, The*, **2013**, 346, f2213	5.9	34
148	Identification of a G-Protein Subunit-α11 Gain-of-Function Mutation, Val340Met, in a Family With Autosomal Dominant Hypocalcemia Type 2 (ADH2). *Journal of Bone and Mineral Research*, **2016**, 31, 1207-14	6.3	33
147	Autosomal dominant hypercalciuria in a mouse model due to a mutation of the epithelial calcium channel, TRPV5. *PLoS ONE*, **2013**, 8, e55412	3.7	32
146	A G-protein Subunit-α11 Loss-of-Function Mutation, Thr54Met, Causes Familial Hypocalciuric Hypercalcemia Type 2 (FHH2). *Journal of Bone and Mineral Research*, **2016**, 31, 1200-6	6.3	32
145	Identification of a second kindred with familial hypocalciuric hypercalcemia type 3 (FHH3) narrows localization to a . *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, 1947-54	5.6	30
144	miR-135b- and miR-146b-dependent silencing of calcium-sensing receptor expression in colorectal tumors. *International Journal of Cancer*, **2016**, 138, 137-45	7.5	30
143	Cinacalcet for Symptomatic Hypercalcemia Caused by AP2S1 Mutations. *New England Journal of Medicine*, **2016**, 374, 1396-1398	59.2	29
142	A missense GATA3 mutation, Thr272Ile, causes the hypoparathyroidism, deafness, and renal dysplasia syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 3897-904	5.6	29
141	MEN1 gene replacement therapy reduces proliferation rates in a mouse model of pituitary adenomas. *Cancer Research*, **2012**, 72, 5060-8	10.1	29
140	Cinacalcet Rectifies Hypercalcemia in a Patient With Familial Hypocalciuric Hypercalcemia Type 2 (FHH2) Caused by a Germline Loss-of-Function G Mutation. *Journal of Bone and Mineral Research*, **2018**, 33, 32-41	6.3	28
139	Mouse models for inherited endocrine and metabolic disorders. *Journal of Endocrinology*, **2011**, 211, 211-30	4.7	28
138	A mouse model for spondyloepiphyseal dysplasia congenita with secondary osteoarthritis due to a Col2a1 mutation. *Journal of Bone and Mineral Research*, **2012**, 27, 413-28	6.3	27

137	Clinical features of X-linked nephrolithiasis in childhood. *Pediatric Nephrology*, **1998**, 12, 625-9	3.2	27
136	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, 265-71	6.5	27
135	Renal chloride channel, CLCN5, mutations in Dent's disease. *Journal of Bone and Mineral Research*, **1999**, 14, 1536-42	6.3	27
134	Allosteric Modulation of the Calcium-sensing Receptor Rectifies Signaling Abnormalities Associated with G-protein 11 Mutations Causing Hypercalcemic and Hypocalcemic Disorders. *Journal of Biological Chemistry*, **2016**, 291, 10876-85	5.4	27
133	Genetic variants of calcium and vitamin D metabolism in kidney stone disease. *Nature Communications*, **2019**, 10, 5175	17.4	27
132	A mouse model for inherited renal fibrosis associated with endoplasmic reticulum stress. *DMM Disease Models and Mechanisms*, **2017**, 10, 773-786	4.1	26
131	A calcium-sensing receptor mutation causing hypocalcemia disrupts a transmembrane salt bridge to activate arrestin-biased signaling. *Science Signaling*, **2018**, 11,	8.8	26
130	Mutational analysis of CLC-5, cofilin and CLC-4 in patients with Dent's disease. *Nephron Physiology*, **2009**, 112, p53-62		26
129	Construction of a 1.2-Mb sequence-ready contig of chromosome 11q13 encompassing the multiple endocrine neoplasia type 1 (MEN1) gene. The European Consortium on MEN1. *Genomics*, **1997**, 44, 94-100	4.3	26
128	Comparative ontogeny, processing, and segmental distribution of the renal chloride channel, ClC-5. *Kidney International*, **2004**, 65, 198-208	9.9	26
127	G mutation in mice causes hypocalcemia rectifiable by calcilytic therapy. *JCI Insight*, **2017**, 2, e91103	9.9	25
126	Genetic mapping studies of familial juvenile hyperuricemic nephropathy on chromosome 16p11-p13. *Journal of Clinical Endocrinology and Metabolism*, **2003**, 88, 464-70	5.6	24
125	Knockin mouse with mutant G mimics human inherited hypocalcemia and is rescued by pharmacologic inhibitors. *JCI Insight*, **2017**, 2, e91079	9.9	23
124	GATA3 mutations found in breast cancers may be associated with aberrant nuclear localization, reduced transactivation and cell invasiveness. *Hormones and Cancer*, **2013**, 4, 123-39	5	23
123	An N-ethyl-N-nitrosourea induced corticotropin-releasing hormone promoter mutation provides a mouse model for endogenous glucocorticoid excess. *Endocrinology*, **2014**, 155, 908-22	4.8	23
122	International Union of Basic and Clinical Pharmacology. CVIII. Calcium-Sensing Receptor Nomenclature, Pharmacology, and Function. *Pharmacological Reviews*, **2020**, 72, 558-604	22.5	22
121	Pasireotide Therapy of Multiple Endocrine Neoplasia Type 1-Associated Neuroendocrine Tumors in Female Mice Deleted for an Men1 Allele Improves Survival and Reduces Tumor Progression. *Endocrinology*, **2016**, 157, 1789-98	4.8	22
120	Confusing genes: a patient with MEN2A and Cushing's disease. *Clinical Endocrinology*, **2013**, 78, 966-8	3.4	22

119	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	11	21
118	Mutant Mice With Calcium-Sensing Receptor Activation Have Hyperglycemia That Is Rectified by Calcilytic Therapy. *Endocrinology*, **2017**, 158, 2486-2502	4.8	21
117	Epidemiology of uromodulin-associated kidney disease - results from a nation-wide survey. *Nephron Extra*, **2012**, 2, 147-58		21
116	SEDLIN forms homodimers: characterisation of SEDLIN mutations and their interactions with transcription factors MBP1, PITX1 and SF1. *PLoS ONE*, **2010**, 5, e10646	3.7	21
115	Multiple Endocrine Neoplasia Type 1: Latest Insights. *Endocrine Reviews*, **2021**, 42, 133-170	27.2	21
114	Calcimimetic and calcilytic therapies for inherited disorders of the calcium-sensing receptor signalling pathway. *British Journal of Pharmacology*, **2018**, 175, 4083-4094	8.6	21
113	Genome-wide study of familial juvenile hyperuricaemic (gouty) nephropathy (FJHN) indicates a new locus, FJHN3, linked to chromosome 2p22.1-p21. *Human Genetics*, **2011**, 129, 51-8	6.3	20
112	Characteristics of hearing loss in HDR (hypoparathyroidism, sensorineural deafness, renal dysplasia) syndrome. *Audiology and Neuro-Otology*, **2006**, 11, 373-9	2.2	20
111	Absence of mutations in the growth hormone (GH)-releasing hormone receptor gene in GH-secreting pituitary adenomas. *Clinical Endocrinology*, **2001**, 54, 301-7	3.4	20
110	Sequence analysis of 139 kb in Xp22.1 containing spermine synthase and the 5' region of PEX. *Genomics*, **1997**, 44, 227-31	4.3	19
109	Mutational analysis in X-linked spondyloepiphyseal dysplasia tarda. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 3233-6	5.6	19
108	Genetics of kidney stone disease. *Nature Reviews Urology*, **2020**, 17, 407-421	5.5	18
107	Expression and chromosomal localization of the Requiem gene. *Mammalian Genome*, **1998**, 9, 660-5	3.2	18
106	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	5.6	17
105	Role of Ca2+ and L-Phe in regulating functional cooperativity of disease-associated "toggle" calcium-sensing receptor mutations. *PLoS ONE*, **2014**, 9, e113622	3.7	17
104	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, E1300-5	5.6	17
103	A novel EXT1 splice site mutation in a kindred with hereditary multiple exostosis and osteoporosis. *Journal of Clinical Endocrinology and Metabolism*, **2005**, 90, 5386-92	5.6	17
102	A five-base pair deletion in the sedlin gene causes spondyloepiphyseal dysplasia tarda in a six-generation Arkansas kindred. *Journal of Clinical Endocrinology and Metabolism*, **2000**, 85, 3343-7	5.6	17

101	Multiple Endocrine Neoplasia Type 1 **2010**, 2719-2741		17
100	X-linked hypoparathyroidism region on Xq27 is evolutionarily conserved with regions on 3q26 and 13q34 and contains a novel P-type ATPase. *Genomics*, **2004**, 84, 1060-70	4.3	16
99	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 - An International Consensus Statement. *Neuroendocrinology*, **2021**, 111, 609-630	5.6	16
98	A mouse with an N-Ethyl-N-nitrosourea (ENU) Induced Trp589Arg Galnt3 mutation represents a model for hyperphosphataemic familial tumoural calcinosis. *PLoS ONE*, **2012**, 7, e43205	3.7	15
97	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	3.7	15
96	Molecular Genetic Studies of Pancreatic Neuroendocrine Tumors: New Therapeutic Approaches. *Endocrinology and Metabolism Clinics of North America*, **2018**, 47, 525-548	5.5	14
95	A familial syndrome due to Arg648Stop mutation in the X-linked renal chloride channel gene. *Pediatric Nephrology*, **1999**, 13, 278-83	3.2	14
94	Construction of a YAC contig and an STS map spanning 3.6 megabase pairs in Xp22.1. *Human Genetics*, **1996**, 97, 60-8	6.3	14
93	Hormonal regulation of biomineralization. *Nature Reviews Endocrinology*, **2021**, 17, 261-275	15.2	14
92	Asymmetric activation of the calcium-sensing receptor homodimer. *Nature*, **2021**, 595, 455-459	50.4	14
91	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	3.7	13
90	Proliferation rates of multiple endocrine neoplasia type 1 (MEN1)-associated tumors. *Endocrinology* , **2012**, 153, 5167-79	4.8	13
89	A novel MEN1 intronic mutation associated with multiple endocrine neoplasia type 1. *Clinical Endocrinology*, **2007**, 66, 709-13	3.4	12
88	Association of prolactin receptor (PRLR) variants with prolactinomas. *Human Molecular Genetics*, **2019**, 28, 1023-1037	5.6	12
87	Medial Arterial Calcification: JACC State-of-the-Art Review. *Journal of the American College of Cardiology*, **2021**, 78, 1145-1165	15.1	12
86	Utility of Population-Level DNA Sequence Data in the Diagnosis of Hereditary Endocrine Disease. *Journal of the Endocrine Society*, **2017**, 1, 1507-1526	0.4	11
85	N-ethyl-N-nitrosourea-Induced Adaptor Protein 2 Sigma Subunit 1 () Mutations Establish Loss-of-Function Mice. *JBMR Plus*, **2017**, 1, 3-15	3.9	11
84	Frequent Occurrence of an Intron 4 Mutation in Multiple Endocrine Neoplasia Type 1		11

83	Genetic approaches to metabolic bone diseases. *British Journal of Clinical Pharmacology*, **2019**, 85, 1147-1160	3.8	11
82	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	12.7	10
81	Hypoparathyroidism, deafness, and renal dysplasia syndrome: 20 Years after the identification of the first GATA3 mutations. *Human Mutation*, **2020**, 41, 1341-1350	4.7	10
80	Large-scale exome datasets reveal a new class of adaptor-related protein complex 2 sigma subunit (AP2σ) mutations, located at the interface with the AP2 alpha subunit, that impair calcium-sensing receptor signalling. *Human Molecular Genetics*, **2018**, 27, 901-911	5.6	10
79	Linkage studies of a Missouri kindred with autosomal dominant spondyloepimetaphyseal dysplasia (SEMD) indicate genetic heterogeneity. *Journal of Bone and Mineral Research*, **1997**, 12, 1204-9	6.3	10
78	Cinacalcet corrects hypercalcemia in mice with an inactivating Gα11 mutation. *JCI Insight*, **2017**, 2,	9.9	10
77	Regulation of sclerostin in glucocorticoid-induced osteoporosis (GIO) in mice and humans. *Endocrine Connections*, **2019**, 8, 923-934	3.5	10
76	Clinical MEN-1 Among a Large Cohort of Patients With Acromegaly. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	10
75	The calcium-sensing receptor: And its involvement in parathyroid pathology. *Annales D'Endocrinologie*, **2015**, 76, 81-3	1.7	9
74	Mutant prolactin receptor and familial hyperprolactinemia. *New England Journal of Medicine*, **2014**, 370, 977-8	59.2	9
73	The Role of Molecular Genetics in Screening for Multiple Endocrine Neoplasia Type 1. *Endocrinology and Metabolism Clinics of North America*, **1994**, 23, 117-135	5.5	9
72	Mice with an N-Ethyl-N-Nitrosourea (ENU) Induced Tyr209Asn Mutation in Natriuretic Peptide Receptor 3 (NPR3) Provide a Model for Kyphosis Associated with Activation of the MAPK Signaling Pathway. *PLoS ONE*, **2016**, 11, e0167916	3.7	9
71	MiR-15a/miR-16-1 expression inversely correlates with cyclin D1 levels in Men1 pituitary NETs. *Journal of Endocrinology*, **2018**,	4.7	9
70	Somatic mutations of GNA11 and GNAQ in CTNNB1-mutant aldosterone-producing adenomas presenting in puberty, pregnancy or menopause. *Nature Genetics*, **2021**, 53, 1360-1372	36.3	9
69	What is the appropriate management of nonfunctioning pancreatic neuroendocrine tumours disclosed on screening in adult patients with multiple endocrine neoplasia type 1?. *Clinical Endocrinology*, **2019**, 91, 708-715	3.4	8
68	Comparison of human chromosome 19q13 and syntenic region on mouse chromosome 7 reveals absence, in man, of 11.6 Mb containing four mouse calcium-sensing receptor-related sequences: relevance to familial benign hypocalciuric hypercalcaemia type 3. *European Journal of Human Genetics*, **2010**, 18, 442-7	5.3	8
67	1. Multiple Endocrine Neoplasia Type 1. *Translational Endocrinology & Metabolism*, **2011**, 13-44		8
66	Rickets and osteomalacia. *Medicine*, **2009**, 37, 483-488	0.6	8

65	The role of biomineralization in disorders of skeletal development and tooth formation. *Nature Reviews Endocrinology*, **2021**, 17, 336-349	15.2	8
64	Molecular genetics of disorders of calcium homeostasis. *Baillieres Clinical Endocrinology and Metabolism*, **1995**, 9, 581-608		7
63	EagI and NotI linking clones from human chromosomes 11 and Xp. *Human Genetics*, **1996**, 97, 742-9	6.3	7
62	Molecular genetics of parathyroid disease. *Current Opinion in Endocrinology, Diabetes and Obesity*, **1996**, 3, 521-528		7
61	Familial and Hereditary Forms of Primary Hyperparathyroidism **2015**, 341-363		6
60	A mouse model of early-onset renal failure due to a xanthine dehydrogenase nonsense mutation. *PLoS ONE*, **2012**, 7, e45217	3.7	6
59	Kidney stones: a fetal origins hypothesis. *Journal of Bone and Mineral Research*, **2013**, 28, 2535-9	6.3	6
58	Calcium Regulation, Calcium Homeostasis, and Genetic Disorders of Calcium Metabolism **2010**, 1136-1159		6
57	Hypocalcaemic disorders, hypoparathyroidism, and pseudohypoparathyroidism **2011**, 675-686		6
56	Age-dependent changes in protein incorporation into collagen-rich tissues of mice by in vivo pulsed SILAC labelling. *ELife*, **2021**, 10,	8.9	6
55	Multiple Endocrine Neoplasia Type 1 **2016**, 2566-2593.e9		5
54	Molecular genetic advances in pituitary tumor development. *Expert Review of Endocrinology and Metabolism*, **2015**, 10, 35-53	4.1	5
53	Cox-2 promotes chromogranin A expression and bioactivity: evidence for a prostaglandin E2-dependent mechanism and the involvement of a proximal cyclic adenosine 5'-monophosphate-responsive element. *Endocrinology*, **2007**, 148, 4310-7	4.8	5
52	Bone Mineral Content and Density. *Current Protocols in Mouse Biology*, **2012**, 2, 365-400	1.1	5
51	Regulation of Calcium Homeostasis and Genetic Disorders that Affect Calcium Metabolism **2016**, 1063-1089.e10		4
50	MANAGEMENT OF ENDOCRINE DISEASE: Postsurgical hypoparathyroidism: current treatments and future prospects for parathyroid allotransplantation. *European Journal of Endocrinology*, **2021**, 184, R165-R175	6.5	4
49	Pseudohypoparathyroidism type 1a due to a novel mutation in the GNAS gene. *Clinical Endocrinology*, **2016**, 84, 463-5	3.4	4
48	Mice with a Brd4 Mutation Represent a New Model of Nephrocalcinosis. *Journal of Bone and Mineral Research*, **2019**, 34, 1324-1335	6.3	3

47	Rickets and osteomalacia. *Medicine*, **2013**, 41, 594-599	0.6	3
46	Fragmentation of filtered proteins and implications for glomerular protein sieving in Fanconi syndrome. *Kidney International*, **2002**, 62, 349	9.9	3
45	Dent's disease. *Nephrology Dialysis Transplantation*, **2005**, 20, 2284-5	4.3	3
44	Multiple Endocrine Neoplasia Type 1 (MEN1) Phenocopy Due to a Cell Cycle Division 73 () Variant. *Journal of the Endocrine Society*, **2020**, 4, bvaa142	0.4	3
43	Studies of mice deleted for Sox3 and uc482: relevance to X-linked hypoparathyroidism. *Endocrine Connections*, **2020**,	3.5	3
42	Parathyroid Disorders **2003**, 485-508		3
41	Activating Mutations of the G-protein Subunit 11 Interdomain Interface Cause Autosomal Dominant Hypocalcemia Type 2. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	3
40	Multiple Endocrine Neoplasia Type 1 (MEN1) 5'UTR Deletion, in MEN1 Family, Decreases Menin Expression. *Journal of Bone and Mineral Research*, **2021**, 36, 100-109	6.3	3
39	Neonatal Hypocalcemic Seizures in Offspring of a Mother With Familial Hypocalciuric Hypercalcemia Type 1 (FHH1). *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	2
38	Introduction to Genetics of Skeletal and Mineral Metabolic Diseases **2018**, 1-21		2
37	Multiple endocrine neoplasia. *Medicine*, **2013**, 41, 562-565	0.6	2
36	Clinically relevant genetic advances in endocrinology. *Clinical Medicine*, **2013**, 13, 299-305	1.9	2
35	Hypoparathyroidism **2013**, 409-423		2
34	Control of PTH secretion by the TRPC1 ion channel. *JCI Insight*, **2020**, 5,	9.9	2
33	Genetic background influences tumour development in heterozygous Men1 knockout mice. *Endocrine Connections*, **2020**, 9, 426-437	3.5	2
32	Aberrant methylation underlies insulin gene expression in human insulinoma. *Nature Communications*, **2020**, 11, 5210	17.4	2
31	Ap2s1 mutation causes hypercalcaemia in mice and impairs interaction between calcium-sensing receptor and adaptor protein-2. *Human Molecular Genetics*, **2021**, 30, 880-892	5.6	2
30	Multiple endocrine neoplasia type 1 in children and adolescents: Clinical features and treatment outcomes. *Surgery*, **2021**,	3.6	2

29	An N-Ethyl-N-Nitrosourea (ENU)-Induced Tyr265Stop Mutation of the DNA Polymerase Accessory Subunit Gamma 2 (Polg2) Is Associated With Renal Calcification in Mice. *Journal of Bone and Mineral Research*, **2019**, 34, 497-507	6.3	2
28	An -Ethyl--Nitrosourea (ENU) Mutagenized Mouse Model for Autosomal Dominant Nonsyndromic Kyphoscoliosis Due to Vertebral Fusion. *JBMR Plus*, **2018**, 2, 154-163	3.9	1
27	Mouse Models: Approaches to Generating in vivo Models for Hereditary Disorders of Mineral and Skeletal Homeostasis **2013**, 181-204		1
26	X-Linked Nephrolithiasis/Dent Disease and Mutations in the ClC-5 Chloride Channel **2000**, 133-152		1
25	Multiple Endocrine Neoplasia Type 1 **2006**, 386-392		1
24	Genetic Disorders of Calcium and Phosphate Homeostasis **2009**, 267-305		1
23	Metabolic Bone Disease in Children **1998**, 759-783		1
22	Whole genome sequence analysis identifies a PAX2 mutation to establish a correct diagnosis for a syndromic form of hyperuricemia. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2521-2528	2.5	1
21	Calcilytic NPSP795 Increases Plasma Calcium and PTH in an Autosomal Dominant Hypocalcemia Type 1 Mouse Model. *JBMR Plus*, **2020**, 4, e10402	3.9	1
20	PTH Infusion for Seizures in Autosomal Dominant Hypocalcemia Type 1. *New England Journal of Medicine*, **2021**, 385, 189-191	59.2	1
19	Genetics of Skeletal Disorders. *Handbook of Experimental Pharmacology*, **2020**, 262, 325-351	3.2	0
18	Genetic Disorders of Calcium and Phosphate Homeostasis **1996**, 311-345		0
17	Familial States of Primary Hyperparathyroidism **2018**, 629-638		0
16	Autosomal Dominant Hypocalcemia Type 1 (ADH1) Associated With Myoclonus and Intracerebral Calcifications.. *Journal of the Endocrine Society*, **2022**, 6, bvac042	0.4	0
15	Mouse Models: Approaches to Generate In Vivo Models for Hereditary Disorders of Mineral and Skeletal Homeostasis **2018**, 89-118		
14	Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis **2014**, 1-59		
13	Hypoparathyroidism and Pseudohypoparathyroidism **2012**, 273-288		
12	Parathyroid Disorders **2012**, 557-588		

11	Multiple Endocrine Neoplasia Type 1 **2013**, 479-504	
10	Multiple endocrine neoplasia. *Medicine*, **2009**, 37, 450-453	0.6
9	Chloride channel mutations in hypercalciuric kidney stone disease. *Clinical and Experimental Nephrology*, **1998**, 2, 194-198	2.5
8	Rickets and Osteomalacia. *Medicine*, **2001**, 29, 74-80	0.6
7	Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis **2016**, 291-339	
6	Genetic Basis of Renal Stones884-892	
5	Genetic regulation of parathyroid gland development **2020**, 1355-1377	
4	Small molecules restore the function of mutant CLC5 associated with Dent disease. *Journal of Cellular and Molecular Medicine*, **2021**, 25, 1319-1322	5.6
3	Hypoparathyroidism **2018**, 617-636	
2	Multiple Endocrine Neoplasia Syndromes **2018**, 699-732	
1	The bromodomain inhibitor JQ1+ reduces calcium-sensing receptor activity in pituitary cell lines. *Journal of Molecular Endocrinology*, **2021**, 67, 83-94	4.5