Rajesh V Thakker

List of Publications by Year in Descending Order

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	Autosomal Dominant Hypocalcemia Type 1 (ADH1) Associated With Myoclonus and Intracerebral Calcifications.. *Journal of the Endocrine Society*, **2022**, 6, bvac042	0.4	0
261	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
260	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
259	Hormonal regulation of biomineralization. *Nature Reviews Endocrinology*, **2021**, 17, 261-275	15.2	14
258	The role of biomineralization in disorders of skeletal development and tooth formation. *Nature Reviews Endocrinology*, **2021**, 17, 336-349	15.2	8
257	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
256	Multiple endocrine neoplasia type 1 in children and adolescents: Clinical features and treatment outcomes. *Surgery*, **2021**,	3.6	2
255	Asymmetric activation of the calcium-sensing receptor homodimer. *Nature*, **2021**, 595, 455-459	50.4	14
254	Multiple Endocrine Neoplasia Type 1: Latest Insights. *Endocrine Reviews*, **2021**, 42, 133-170	27.2	21
253	Small molecules restore the function of mutant CLC5 associated with Dent disease. *Journal of Cellular and Molecular Medicine*, **2021**, 25, 1319-1322	5.6	
252	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
251	PTH Infusion for Seizures in Autosomal Dominant Hypocalcemia Type 1. *New England Journal of Medicine*, **2021**, 385, 189-191	59.2	1
250	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
249	Age-dependent changes in protein incorporation into collagen-rich tissues of mice by in vivo pulsed SILAC labelling. *ELife*, **2021**, 10,	8.9	6
248	Medial Arterial Calcification: JACC State-of-the-Art Review. *Journal of the American College of Cardiology*, **2021**, 78, 1145-1165	15.1	12
247	The bromodomain inhibitor JQ1+ reduces calcium-sensing receptor activity in pituitary cell lines. *Journal of Molecular Endocrinology*, **2021**, 67, 83-94	4.5	
246	International Union of Basic and Clinical Pharmacology. CVIII. Calcium-Sensing Receptor Nomenclature, Pharmacology, and Function. *Pharmacological Reviews*, **2020**, 72, 558-604	22.5	22

245	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
244	Genetics of kidney stone disease. *Nature Reviews Urology*, **2020**, 17, 407-421	5.5	18
243	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
242	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
241	Genetics of Skeletal Disorders. *Handbook of Experimental Pharmacology*, **2020**, 262, 325-351	3.2	0
240	Control of PTH secretion by the TRPC1 ion channel. *JCI Insight*, **2020**, 5,	9.9	2
239	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
238	Studies of mice deleted for Sox3 and uc482: relevance to X-linked hypoparathyroidism. *Endocrine Connections*, **2020**,	3.5	3
237	Genetic background influences tumour development in heterozygous Men1 knockout mice. *Endocrine Connections*, **2020**, 9, 426-437	3.5	2
236	Genetic regulation of parathyroid gland development **2020**, 1355-1377		
235	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
234	Aberrant methylation underlies insulin gene expression in human insulinoma. *Nature Communications*, **2020**, 11, 5210	17.4	2
233	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
232	Calcilytic NPSP795 Increases Plasma Calcium and PTH in an Autosomal Dominant Hypocalcemia Type 1 Mouse Model. *JBMR Plus*, **2020**, 4, e10402	3.9	1
231	Clinical MEN-1 Among a Large Cohort of Patients With Acromegaly. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	10
230	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
229	Mice with a Brd4 Mutation Represent a New Model of Nephrocalcinosis. *Journal of Bone and Mineral Research*, **2019**, 34, 1324-1335	6.3	3
228	MANAGEMENT OF ENDOCRINE DISEASE: Unmet therapeutic, educational and scientific needs in parathyroid disorders. *European Journal of Endocrinology*, **2019**, 181, P1-P19	6.5	40

227	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
226	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
225	Regulation of sclerostin in glucocorticoid-induced osteoporosis (GIO) in mice and humans. *Endocrine Connections*, **2019**, 8, 923-934	3.5	10
224	Standards of care for hypoparathyroidism in adults: a Canadian and International Consensus. *European Journal of Endocrinology*, **2019**, 180, P1-P22	6.5	44
223	Genetic variants of calcium and vitamin D metabolism in kidney stone disease. *Nature Communications*, **2019**, 10, 5175	17.4	27
222	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
221	Association of prolactin receptor (PRLR) variants with prolactinomas. *Human Molecular Genetics*, **2019**, 28, 1023-1037	5.6	12
220	Genetic approaches to metabolic bone diseases. *British Journal of Clinical Pharmacology*, **2019**, 85, 1147-1160	3.8	11
219	A calcium-sensing receptor mutation causing hypocalcemia disrupts a transmembrane salt bridge to activate -arrestin-biased signaling. *Science Signaling*, **2018**, 11,	8.8	26
218	Current and emerging therapies for PNETs in patients with or without MEN1. *Nature Reviews Endocrinology*, **2018**, 14, 216-227	15.2	34
217	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
216	Large-scale exome datasets reveal a new class of adaptor-related protein complex 2 sigma subunit (AP2 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
215	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
214	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
213	Introduction to Genetics of Skeletal and Mineral Metabolic Diseases **2018**, 1-21		2
212	Mouse Models: Approaches to Generate In Vivo Models for Hereditary Disorders of Mineral and Skeletal Homeostasis **2018**, 89-118		
211	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
210	Molecular Genetic Studies of Pancreatic Neuroendocrine Tumors: New Therapeutic Approaches. *Endocrinology and Metabolism Clinics of North America*, **2018**, 47, 525-548	5.5	14

209	MiR-15a/miR-16-1 expression inversely correlates with cyclin D1 levels in Men1 pituitary NETs. *Journal of Endocrinology*, **2018**,	4.7	9
208	The calcium-sensing receptor in physiology and in calcitropic and noncalcitropic diseases. *Nature Reviews Endocrinology*, **2018**, 15, 33-51	15.2	113
207	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
206	Familial States of Primary Hyperparathyroidism **2018**, 629-638		0
205	Hypoparathyroidism **2018**, 617-636		
204	Multiple Endocrine Neoplasia Syndromes **2018**, 699-732		
203	A mouse model for inherited renal fibrosis associated with endoplasmic reticulum stress. *DMM Disease Models and Mechanisms*, **2017**, 10, 773-786	4.1	26
202	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
201	Knockin mouse with mutant G mimics human inherited hypocalcemia and is rescued by pharmacologic inhibitors. *JCI Insight*, **2017**, 2, e91079	9.9	23
200	Hypoparathyroidism. *Nature Reviews Disease Primers*, **2017**, 3, 17055	51.1	100
199	Molecular genetics of syndromic and non-syndromic forms of parathyroid carcinoma. *Human Mutation*, **2017**, 38, 1621-1648	4.7	55
198	Hypercalcemic Disorders in Children. *Journal of Bone and Mineral Research*, **2017**, 32, 2157-2170	6.3	51
197	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
196	Mutant Mice With Calcium-Sensing Receptor Activation Have Hyperglycemia That Is Rectified by Calcilytic Therapy. *Endocrinology*, **2017**, 158, 2486-2502	4.8	21
195	G mutation in mice causes hypocalcemia rectifiable by calcilytic therapy. *JCI Insight*, **2017**, 2, e91103	9.9	25
194	Genomic profiling reveals mutational landscape in parathyroid carcinomas. *JCI Insight*, **2017**, 2, e92061	9.9	50
193	Cinacalcet corrects hypercalcemia in mice with an inactivating G1 mutation. *JCI Insight*, **2017**, 2,	9.9	10
192	Multiple Endocrine Neoplasia Type 1 **2016**, 2566-2593.e9		5

191	Regulation of Calcium Homeostasis and Genetic Disorders that Affect Calcium Metabolism **2016**, 1063-1089.e10	4	
190	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
189	Cinacalcet for Symptomatic Hypercalcemia Caused by AP2S1 Mutations. *New England Journal of Medicine*, **2016**, 374, 1396-1398	59.2	29
188	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
187	Management of Hypoparathyroidism: Summary Statement and Guidelines. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 2273-83	5.6	207
186	Presentation of Hypoparathyroidism: Etiologies and Clinical Features. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 2300-12	5.6	175
185	Epidemiology and Diagnosis of Hypoparathyroidism. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 2284-99	5.6	148
184	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
183	Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis **2016**, 291-339		
182	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
181	Allosteric Modulation of the Calcium-sensing Receptor Rectifies Signaling Abnormalities Associated with G-protein α11 Mutations Causing Hypercalcemic and Hypocalcemic Disorders. *Journal of Biological Chemistry*, **2016**, 291, 10876-85	5.4	27
180	Pseudohypoparathyroidism type 1a due to a novel mutation in the GNAS gene. *Clinical Endocrinology*, **2016**, 84, 463-5	3.4	4
179	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
178	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
177	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
176	The calcium-sensing receptor: And its involvement in parathyroid pathology. *Annales D'Endocrinologie*, **2015**, 76, 81-3	1.7	9
175	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
174	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

173	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. *Nature Genetics*, **2015**, 47, 717-726	36.3	244
172	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
171	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
170	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
169	Familial and Hereditary Forms of Primary Hyperparathyroidism **2015**, 341-363		6
168	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
167	GNAS mutations in Pseudohypoparathyroidism type 1a and related disorders. *Human Mutation*, **2015**, 36, 11-9	4.7	82
166	Molecular genetic advances in pituitary tumor development. *Expert Review of Endocrinology and Metabolism*, **2015**, 10, 35-53	4.1	5
165	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
164	Multiple endocrine neoplasia type 1 (MEN1) and type 4 (MEN4). *Molecular and Cellular Endocrinology*, **2014**, 386, 2-15	4.4	261
163	Mutant prolactin receptor and familial hyperprolactinemia. *New England Journal of Medicine*, **2014**, 370, 977-8	59.2	9
162	Diagnosis of asymptomatic primary hyperparathyroidism: proceedings of the Fourth International Workshop. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, 3570-9	5.6	233
161	Guidelines for the management of thyroid cancer. *Clinical Endocrinology*, **2014**, 81 Suppl 1, 1-122	3.4	692
160	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
159	CDC73 intragenic deletion in familial primary hyperparathyroidism associated with parathyroid carcinoma. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, 3044-8	5.6	39
158	Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis **2014**, 1-59		
157	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
156	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

155	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
154	Mutant prolactin receptor and familial hyperprolactinemia. *New England Journal of Medicine*, **2013**, 369, 2012-2020	59.2	80
153	Confusing genes: a patient with MEN2A and Cushing's disease. *Clinical Endocrinology*, **2013**, 78, 966-8	3.4	22
152	Rickets and osteomalacia. *Medicine*, **2013**, 41, 594-599	0.6	3
151	Multiple endocrine neoplasia. *Medicine*, **2013**, 41, 562-565	0.6	2
150	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
149	Investigating hypocalcaemia. *BMJ, The*, **2013**, 346, f2213	5.9	34
148	Mutations affecting G-protein subunit 11 in hypercalcemia and hypocalcemia. *New England Journal of Medicine*, **2013**, 368, 2476-2486	59.2	282
147	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
146	Clinically relevant genetic advances in endocrinology. *Clinical Medicine*, **2013**, 13, 299-305	1.9	2
145	Whole-exome sequencing studies of nonfunctioning pituitary adenomas. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, E796-800	5.6	69
144	Kidney stones: a fetal origins hypothesis. *Journal of Bone and Mineral Research*, **2013**, 28, 2535-9	6.3	6
143	Hypoparathyroidism **2013**, 409-423		2
142	Multiple Endocrine Neoplasia Type 1 **2013**, 479-504		
141	Mouse Models: Approaches to Generating in vivo Models for Hereditary Disorders of Mineral and Skeletal Homeostasis **2013**, 181-204		1
140	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
139	Mutations in AP2S1 cause familial hypocalciuric hypercalcemia type 3. *Nature Genetics*, **2013**, 45, 93-7	36.3	196
138	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

137	Hypoparathyroidism and Pseudohypoparathyroidism **2012**, 273-288		
136	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
135	A mouse model of early-onset renal failure due to a xanthine dehydrogenase nonsense mutation. *PLoS ONE*, **2012**, 7, e45217	3.7	6
134	Epidemiology of uromodulin-associated kidney disease - results from a nation-wide survey. *Nephron Extra*, **2012**, 2, 147-58		21
133	Parathyroid Disorders **2012**, 557-588		
132	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
131	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
130	Clinical practice guidelines for multiple endocrine neoplasia type 1 (MEN1). *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, 2990-3011	5.6	830
129	MEN1 gene replacement therapy reduces proliferation rates in a mouse model of pituitary adenomas. *Cancer Research*, **2012**, 72, 5060-8	10.1	29
128	Whole-exome sequencing studies of nonhereditary (sporadic) parathyroid adenomas. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E1995-2005	5.6	101
127	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
126	Proliferation rates of multiple endocrine neoplasia type 1 (MEN1)-associated tumors. *Endocrinology* , **2012**, 153, 5167-79	4.8	13
125	Bone Mineral Content and Density. *Current Protocols in Mouse Biology*, **2012**, 2, 365-400	1.1	5
124	Role of multiple endocrine neoplasia type 1 mutational analysis in clinical practice. *Endocrine Practice*, **2011**, 17 Suppl 3, 8-17	3.2	56
123	Transcription factors in parathyroid development: lessons from hypoparathyroid disorders. *Annals of the New York Academy of Sciences*, **2011**, 1237, 24-38	6.5	42
122	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
121	1. Multiple Endocrine Neoplasia Type 1. *Translational Endocrinology & Metabolism*, **2011**, 13-44		8
120	Mouse models for inherited endocrine and metabolic disorders. *Journal of Endocrinology*, **2011**, 211, 211-30	4.7	28

119	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
118	Hypocalcaemic disorders, hypoparathyroidism, and pseudohypoparathyroidism **2011**, 675-686		6
117	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
116	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
115	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
114	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
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, 2028-38	5.6	44
112	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
111	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
110	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
109	Dent's disease. *Orphanet Journal of Rare Diseases*, **2010**, 5, 28	4.2	133
108	Multiple endocrine neoplasia type 1 (MEN1). *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2010**, 24, 355-70	6.5	161
107	Diagnostic challenges due to phenocopies: lessons from Multiple Endocrine Neoplasia type1 (MEN1). *Human Mutation*, **2010**, 31, E1089-101	4.7	53
106	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
105	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
104	Calcium Regulation, Calcium Homeostasis, and Genetic Disorders of Calcium Metabolism **2010**, 1136-1159		6
103	Multiple Endocrine Neoplasia Type 1 **2010**, 2719-2741		17
102	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

101	OCRL1 mutations in Dent 2 patients suggest a mechanism for phenotypic variability. *Nephron Physiology*, **2009**, 112, p27-36		62
100	Mutational analysis of CLC-5, cofilin and CLC-4 in patients with Dent's disease. *Nephron Physiology*, **2009**, 112, p53-62		26
99	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
98	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
97	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
96	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
95	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
94	Rickets and osteomalacia. *Medicine*, **2009**, 37, 483-488	0.6	8
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