# Rajesh V Thakker

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3083896/publications.pdf

Version: 2024-02-01

| | | 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 | Autosomal Dominant Hypocalcemia Type 1 (ADH1) Associated With Myoclonus and Intracerebral Calcifications. Journal of the Endocrine Society, 2022, 6, bvac042. | 0.1 | 5 |
| 2 | Genetics of monogenic disorders of calcium and bone metabolism. Clinical Endocrinology, 2022, 97, 483-501. | 1.2 | 7 |
| 3 | The Bartter-Gitelman Spectrum: 50-Year Follow-up With Revision of Diagnosis After Whole-Genome Sequencing. Journal of the Endocrine Society, 2022, 6, . | 0.1 | 7 |
| 4 | Skeletal and extraskeletal disorders of biomineralization. Nature Reviews Endocrinology, 2022, 18, 473-489. | 4.3 | 25 |
| 5 | Whole Exome Sequencing in Two <scp>Southeast</scp> Asian Families With Atypical Femur Fractures. JBMR Plus, 2022, 6, . | 1.3 | 3 |
| 6 | 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 |
| 7 | Multiple Endocrine Neoplasia Type 1: Latest Insights. Endocrine Reviews, 2021, 42, 133-170. | 8.9 | 85 |
| 8 | Small molecules restore the function of mutant CLC5 associated with Dent disease. Journal of Cellular and Molecular Medicine, 2021, 25, 1319-1322. | 1.6 | 5 |
| 9 | Ap2s1 mutation causes hypercalcaemia in mice and impairs interaction between calcium-sensing receptor and adaptor protein-2. Human Molecular Genetics, 2021, 30, 880-892. | 1.4 | 10 |
| 10 | Hormonal regulation of biomineralization. Nature Reviews Endocrinology, 2021, 17, 261-275. | 4.3 | 50 |
| 11 | The role of biomineralization in disorders of skeletal development and tooth formation. Nature Reviews Endocrinology, 2021, 17, 336-349. | 4.3 | 46 |
| 12 | MANAGEMENT OF ENDOCRINE DISEASE: Postsurgical hypoparathyroidism: current treatments and future prospects for parathyroid allotransplantation. European Journal of Endocrinology, 2021, 184, R165-R175. | 1.9 | 15 |
| 13 | Multiple endocrine neoplasia type 1 in children and adolescents: Clinical features and treatment outcomes. Surgery, 2021, , . | 1.0 | 10 |
| 14 | Asymmetric activation of the calcium-sensing receptor homodimer. Nature, 2021, 595, 455-459. | 13.7 | 59 |
| 15 | PTH Infusion for Seizures in Autosomal Dominant Hypocalcemia Type 1. New England Journal of Medicine, 2021, 385, 189-191. | 13.9 | 11 |
| 16 | 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 |
| 17 | Age-dependent changes in protein incorporation into collagen-rich tissues of mice by in vivo pulsed SILAC labelling. ELife, 2021, 10, . | 2.8 | 22 |
| 18 | Medial Arterial Calcification. Journal of the American College of Cardiology, 2021, 78, 1145-1165. | 1.2 | 106 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | The bromodomain inhibitor JQ1+ reduces calcium-sensing receptor activity in pituitary cell lines. Journal of Molecular Endocrinology, 2021, 67, 83-94. | 1.1 | 1 |
| 20 | Genetic regulation of parathyroid gland development. , 2020, , 1355-1377. | | 0 |
| 21 | Activating Mutations of the G-protein Subunit Î±â€Š11 Interdomain Interface Cause Autosomal Dominant Hypocalcemia Type 2. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 952-963. | 1.8 | 6 |
| 22 | Aberrant methylation underlies insulin gene expression in human insulinoma. Nature Communications, 2020, 11, 5210. | 5.8 | 9 |
| 23 | 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. | 0.7 | 3 |
| 24 | Calcilytic NPSP795 Increases Plasma Calcium and PTH in an Autosomal Dominant Hypocalcemia Type 1 Mouse Model. JBMR Plus, 2020, 4, e10402. | 1.3 | 3 |
| 25 | International Union of Basic and Clinical Pharmacology. CVIII. Calcium-Sensing Receptor Nomenclature, Pharmacology, and Function. Pharmacological Reviews, 2020, 72, 558-604. | 7.1 | 59 |
| 26 | Hypoparathyroidism, deafness, and renal dysplasia syndrome: 20 Years after the identification of the firstGATA3mutations. Human Mutation, 2020, 41, 1341-1350. | 1.1 | 19 |
| 27 | Genetics of kidney stone disease. Nature Reviews Urology, 2020, 17, 407-421. | 1.9 | 81 |
| 28 | 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 |
| 29 | Neonatal Hypocalcemic Seizures in Offspring of a Mother With Familial Hypocalciuric Hypercalcemia Type 1 (FHH1). Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1393-1400. | 1.8 | 7 |
| 30 | Genetics of Skeletal Disorders. Handbook of Experimental Pharmacology, 2020, 262, 325-351. | 0.9 | 3 |
| 31 | Clinical MEN-1 Among a Large Cohort of Patients With Acromegaly. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e2271-e2281. | 1.8 | 19 |
| 32 | Multiple Endocrine Neoplasia Type 1 (MEN1) 5â€²UTR Deletion, in MEN1 Family, Decreases Menin Expression. Journal of Bone and Mineral Research, 2020, 36, 100-109. | 3.1 | 10 |
| 33 | Control of PTH secretion by the TRPC1 ion channel. JCI Insight, 2020, 5, . | 2.3 | 6 |
| 34 | Multiple Endocrine Neoplasia Type 1 (MEN1) Phenocopy Due to a Cell Cycle Division 73 (CDC73) Variant. Journal of the Endocrine Society, 2020, 4, bvaa142. | 0.1 | 5 |
| 35 | Studies of mice deleted for Sox3 and uc482: relevance to X-linked hypoparathyroidism. Endocrine Connections, 2020, 9, 173-186. | 0.8 | 3 |
| 36 | Genetic background influences tumour development in heterozygous Men1 knockout mice. Endocrine Connections, 2020, 9, 426-437. | 0.8 | 5 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | 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. | 1.2 | 14 |
| 40 | 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 |
| 41 | Mice with a Brd4 Mutation Represent a New Model of Nephrocalcinosis. Journal of Bone and Mineral Research, 2019, 34, 1324-1335. | 3.1 | 7 |
| 42 | Standards of care for hypoparathyroidism in adults: a Canadian and International Consensus. European Journal of Endocrinology, 2019, 180, P1-P22. | 1.9 | 81 |
| 43 | Genetic variants of calcium and vitamin D metabolism in kidney stone disease. Nature Communications, 2019, 10, 5175. | 5.8 | 69 |
| 44 | 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. | 3.1 | 3 |
| 45 | Association of prolactin receptor (PRLR) variants with prolactinomas. Human Molecular Genetics, 2019, 28, 1023-1037. | 1.4 | 24 |
| 46 | The calcium-sensing receptor in physiology and in calcitropic and noncalcitropic diseases. Nature Reviews Endocrinology, 2019, 15, 33-51. | 4.3 | 226 |
| 47 | Genetic approaches to metabolic bone diseases. British Journal of Clinical Pharmacology, 2019, 85, 1147-1160. | 1.1 | 21 |
| 48 | Regulation of sclerostin in glucocorticoid-induced osteoporosis (GIO) in mice and humans. Endocrine Connections, 2019, 8, 923-934. | 0.8 | 15 |
| 49 | miR-15a/miR-16-1 expression inversely correlates with cyclin D1 levels in Men1 pituitary NETs. Journal of Endocrinology, 2019, 240, 41-50. | 1.2 | 12 |
| 50 | 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 |
| 51 | Current and emerging therapies for PNETs in patients with or without MEN1. Nature Reviews Endocrinology, 2018, 14, 216-227. | 4.3 | 46 |
| 52 | 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 |
| 53 | Large-scale exome datasets reveal a new class of adaptor-related protein complex 2 sigma subunit (AP2Ïƒ) mutations, located at the interface with the AP2 alpha subunit, that impair calcium-sensing receptor signalling. Human Molecular Genetics, 2018, 27, 901-911. | 1.4 | 15 |
| 54 | An Nâ€Ethylâ€Nâ€Nitrosourea (ENU) Mutagenized Mouse Model for Autosomal Dominant Nonsyndromic Kyphoscoliosis Due to Vertebral Fusion. JBMR Plus, 2018, 2, 154-163. | 1.3 | 1 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | 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 |
| 56 | 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 |
| 57 | Hypoparathyroidism. , 2018, , 617-636. | | 0 |
| 58 | Multiple Endocrine Neoplasia Syndromes. , 2018, , 699-732. | | 0 |
| 59 | Introduction to Genetics of Skeletal and Mineral Metabolic Diseases. , 2018, , 1-21. | | 2 |
| 60 | Mouse Models: Approaches to Generate In Vivo Models for Hereditary Disorders of Mineral and Skeletal Homeostasis. , 2018, , 89-118. | | 0 |
| 61 | Calcium-sensing receptor residues with loss- and gain-of-function mutations are located in regions of conformational change and cause signalling bias. Human Molecular Genetics, 2018, 27, 3720-3733. | 1.4 | 23 |
| 62 | Molecular Genetic Studies of Pancreatic Neuroendocrine Tumors. Endocrinology and Metabolism Clinics of North America, 2018, 47, 525-548. | 1.2 | 17 |
| 63 | Mouse model for inherited renal fibrosis associated with endoplasmic reticulum stress. DMM Disease Models and Mechanisms, 2017, 10, 773-786. | 1.2 | 34 |
| 64 | Hypoparathyroidism. Nature Reviews Disease Primers, 2017, 3, 17055. | 18.1 | 142 |
| 65 | Molecular genetics of syndromic and non-syndromic forms of parathyroid carcinoma. Human Mutation, 2017, 38, 1621-1648. | 1.1 | 82 |
| 66 | Hypercalcemic Disorders in Children. Journal of Bone and Mineral Research, 2017, 32, 2157-2170. | 3.1 | 82 |
| 67 | Nâ€ethylâ€Nâ€nitrosoureaâ€“Induced Adaptor Protein 2 Sigma Subunit 1 (Ap2s1) Mutations Establish Ap2s1 Lossâ€ofâ€Function Mice. JBMR Plus, 2017, 1, 3-15. | 1.3 | 16 |
| 68 | Mutant Mice With Calcium-Sensing Receptor Activation Have Hyperglycemia That Is Rectified by Calcilytic Therapy. Endocrinology, 2017, 158, 2486-2502. | 1.4 | 31 |
| 69 | Utility of Population-Level DNA Sequence Data in the Diagnosis of Hereditary Endocrine Disease. Journal of the Endocrine Society, 2017, 1, 1507-1526. | 0.1 | 15 |
| 70 | Knockin mouse with mutant GÎ±11 mimics human inherited hypocalcemia and is rescued by pharmacologic inhibitors. JCI Insight, 2017, 2, e91079. | 2.3 | 26 |
| 71 | GÎ±11 mutation in mice causes hypocalcemia rectifiable by calcilytic therapy. JCI Insight, 2017, 2, e91103. | 2.3 | 28 |
| 72 | Genomic profiling reveals mutational landscape in parathyroid carcinomas. JCI Insight, 2017, 2, e92061. | 2.3 | 84 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Cinacalcet corrects hypercalcemia in mice with an inactivating GÎ±11 mutation. JCI Insight, 2017, 2, . | 2.3 | 17 |
| 74 | 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 |
| 75 | 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 |
| 76 | Pseudohypoparathyroidism type 1a due to a novel mutation in the <scp>GNAS</scp> gene. Clinical Endocrinology, 2016, 84, 463-465. | 1.2 | 4 |
| 77 | 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 |
| 78 | 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 |
| 79 | 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 |
| 80 | Cinacalcet for Symptomatic Hypercalcemia Caused by AP2S1 Mutations. New England Journal of Medicine, 2016, 374, 1396-1398. | 13.9 | 38 |
| 81 | Pasireotide Therapy of Multiple Endocrine Neoplasia Type 1â€“Associated Neuroendocrine Tumors in Female Mice Deleted for an Men1 Allele Improves Survival and Reduces Tumor Progression. Endocrinology, 2016, 157, 1789-1798. | 1.4 | 26 |
| 82 | Management of Hypoparathyroidism: Summary Statement and Guidelines. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2273-2283. | 1.8 | 303 |
| 83 | Presentation of Hypoparathyroidism: Etiologies and Clinical Features. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2300-2312. | 1.8 | 246 |
| 84 | Epidemiology and Diagnosis of Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2284-2299. | 1.8 | 230 |
| 85 | Multiple Endocrine Neoplasia Type 1. , 2016, , 2566-2593.e9. | | 5 |
| 86 | Regulation of Calcium Homeostasis and Genetic Disorders that Affect Calcium Metabolism. , 2016, , 1063-1089.e10. | | 7 |
| 87 | 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. | 1.1 | 11 |
| 88 | Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis. , 2016, , 291-339. | | 0 |
| 89 | Association Studies of Calcium-Sensing Receptor (CaSR) Polymorphisms with Serum Concentrations of Glucose and Phosphate, and Vascular Calcification in Renal Transplant Recipients. PLoS ONE, 2015, 10, e0119459. | 1.1 | 15 |
| 90 | GNAS Mutations in Pseudohypoparathyroidism Type 1a and Related Disorders. Human Mutation, 2015, 36, 11-19. | 1.1 | 101 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | Molecular genetic advances in pituitary tumor development. Expert Review of Endocrinology and Metabolism, 2015, 10, 35-53. | 1.2 | 5 |
| 92 | 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 |
| 93 | The calcium-sensing receptor: And its involvement in parathyroid pathology. Annales D'Endocrinologie, 2015, 76, 81-83. | 0.6 | 10 |
| 94 | 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 |
| 95 | 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 |
| 96 | Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nature Genetics, 2015, 47, 717-726. | 9.4 | 310 |
| 97 | 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 |
| 98 | 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 |
| 99 | 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 |
| 100 | Familial and Hereditary Forms of Primary Hyperparathyroidism. , 2015, , 341-363. | | 7 |
| 101 | N-ethyl-N-Nitrosourea (ENU) Induced Mutations within the Klotho Gene Lead to Ectopic Calcification and Reduced Lifespan in Mouse Models. PLoS ONE, 2015, 10, e0122650. | 1.1 | 16 |
| 102 | Multiple Endocrine Neoplasia Type 1. , 2015, , 2943-2948. | | 0 |
| 103 | Multiple Endocrine Neoplasia Type 1. , 2015, , 1-6. | | 0 |
| 104 | Role of Ca2+ and L-Phe in Regulating Functional Cooperativity of Disease-Associated â€œToggleâ€• Calcium-Sensing Receptor Mutations. PLoS ONE, 2014, 9, e113622. | 1.1 | 18 |
| 105 | CDC73 Intragenic Deletion in Familial Primary Hyperparathyroidism Associated With Parathyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 3044-3048. | 1.8 | 46 |
| 106 | Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis. , 2014, , 1-59. | | 0 |
| 107 | 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 |
| 108 | Mutational Analysis of the Adaptor Protein 2 Sigma Subunit (AP2S1) Gene: Search for Autosomal Dominant Hypocalcemia Type 3 (ADH3). Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1300-E1305. | 1.8 | 19 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | Multiple endocrine neoplasia type 1 (MEN1) and type 4 (MEN4). Molecular and Cellular Endocrinology, 2014, 386, 2-15. | 1.6 | 334 |
| 110 | Mutant Prolactin Receptor and Familial Hyperprolactinemia. New England Journal of Medicine, 2014, 370, 976-978. | 13.9 | 9 |
| 111 | Diagnosis of Asymptomatic Primary Hyperparathyroidism: Proceedings of the Fourth International Workshop. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 3570-3579. | 1.8 | 296 |
| 112 | Guidelines for the management of thyroid cancer. Clinical Endocrinology, 2014, 81, 1-122. | 1.2 | 961 |
| 113 | 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 |
| 114 | Mutant Prolactin Receptor and Familial Hyperprolactinemia. New England Journal of Medicine, 2013, 369, 2012-2020. | 13.9 | 106 |
| 115 | Confusing genes: a patient with MEN2A and Cushing's disease. Clinical Endocrinology, 2013, 78, 966-968. | 1.2 | 30 |
| 116 | Rickets and osteomalacia. Medicine, 2013, 41, 594-599. | 0.2 | 17 |
| 117 | Multiple endocrine neoplasia. Medicine, 2013, 41, 562-565. | 0.2 | 3 |
| 118 | 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 |
| 119 | Investigating hypocalcaemia. BMJ, The, 2013, 346, f2213-f2213. | 3.0 | 44 |
| 120 | Mutations Affecting G-Protein Subunit Î±<sub>11</sub>in Hypercalcemia and Hypocalcemia. New England Journal of Medicine, 2013, 368, 2476-2486. | 13.9 | 340 |
| 121 | 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 |
| 122 | Clinically relevant genetic advances in endocrinology. Clinical Medicine, 2013, 13, 299-305. | 0.8 | 2 |
| 123 | Whole-Exome Sequencing Studies of Nonfunctioning Pituitary Adenomas. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E796-E800. | 1.8 | 85 |
| 124 | Kidney Stones: A Fetal Origins Hypothesis. Journal of Bone and Mineral Research, 2013, 28, 2535-2539. | 3.1 | 6 |
| 125 | Hypoparathyroidism. , 2013, , 409-423. | | 2 |
| 126 | Multiple Endocrine Neoplasia Type 1. , 2013, , 479-504. | | 0 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Mouse Models. , 2013, , 181-204. | | 1 |
| 128 | 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 |
| 129 | Mutations in AP2S1 cause familial hypocalciuric hypercalcemia type 3. Nature Genetics, 2013, 45, 93-97. | 9.4 | 242 |
| 130 | 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 |
| 131 | MEN1 Gene Replacement Therapy Reduces Proliferation Rates in a Mouse Model of Pituitary Adenomas. Cancer Research, 2012, 72, 5060-5068. | 0.4 | 34 |
| 132 | Whole-Exome Sequencing Studies of Nonhereditary (Sporadic) Parathyroid Adenomas. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1995-E2005. | 1.8 | 121 |
| 133 | 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 |
| 134 | Proliferation Rates of Multiple Endocrine Neoplasia Type 1 (MEN1)-Associated Tumors. Endocrinology, 2012, 153, 5167-5179. | 1.4 | 13 |
| 135 | Hypoparathyroidism and Pseudohypoparathyroidism. , 2012, , 273-288. | | 0 |
| 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. | 1.1 | 19 |
| 137 | A Mouse Model of Early-Onset Renal Failure Due to a Xanthine Dehydrogenase Nonsense Mutation. PLoS ONE, 2012, 7, e45217. | 1.1 | 8 |
| 138 | Epidemiology of Uromodulin-Associated Kidney Disease â€“ Results from a Nation-Wide Survey. Nephron Extra, 2012, 2, 147-158. | 1.1 | 25 |
| 139 | Parathyroid Disorders. , 2012, , 557-588. | | 1 |
| 140 | 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 |
| 141 | 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 |
| 142 | Clinical Practice Guidelines for Multiple Endocrine Neoplasia Type 1 (MEN1). Journal of Clinical Endocrinology and Metabolism, 2012, 97, 2990-3011. | 1.8 | 1,127 |
| 143 | Bone Mineral Content and Density. , 2012, 2, 365-400. | | 9 |
| 144 | 1. Multiple Endocrine Neoplasia Type 1. Translational Endocrinology & Metabolism, 2011, , 13-44. | 0.2 | 8 |

| # | Article | IF | Citations |
|---|---|---|---|
| 145 | Role of Multiple Endocrine Neoplasia Type 1 Mutational Analysis in Clinical Practice. Endocrine Practice, 2011, 17, 8-17. | 1.1 | 71 |
| 146 | Transcription factors in parathyroid development: lessons from hypoparathyroid disorders. Annals of the New York Academy of Sciences, 2011, 1237, 24-38. | 1.8 | 47 |
| 147 | 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, 51-58. | 1.8 | 25 |
| 148 | Mouse models for inherited endocrine and metabolic disorders. Journal of Endocrinology, 2011, 211, 211-230. | 1.2 | 30 |
| 149 | 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 |
| 150 | Hypocalcaemic disorders, hypoparathyroidism, and pseudohypoparathyroidism. , 2011, , 675-686. | | 7 |
| 151 | Multiple Endocrine Neoplasia Type 1. , 2011, , 2394-2398. | | 0 |
| 152 | Diagnostic challenges due to phenocopies: lessons from Multiple Endocrine Neoplasia type1 (MEN1). Human Mutation, 2010, 31, E1089-E1101. | 1.1 | 78 |
| 153 | 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 |
| 154 | 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 |
| 155 | Comparison of human chromosome 19q13 and syntenic region on mouse chromosome 7 reveals absence, in man, of 11.6â€‰Mb containing four mouse calcium-sensing receptor-related sequences: relevance to familial benign hypocalciuric hypercalcaemia type 3. European Journal of Human Genetics, 2010, 18, 442-447. | 1.4 | 8 |
| 156 | SEDLIN Forms Homodimers: Characterisation of SEDLIN Mutations and Their Interactions with Transcription Factors MBP1, PITX1 and SF1. PLoS ONE, 2010, 5, e10646. | 1.1 | 23 |
| 157 | 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 |
| 158 | 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 |
| 159 | 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 |
| 160 | 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 |
| 161 | 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 |
| 162 | Dent's disease. Orphanet Journal of Rare Diseases, 2010, 5, 28. | 1.2 | 181 |

| # | Article | IF | Citations |
|---|---|---|---|
| 163 | Multiple endocrine neoplasia type 1 (MEN1). Best Practice and Research in Clinical Endocrinology and Metabolism, 2010, 24, 355-370. | 2.2 | 206 |
| 164 | Calcium Regulation, Calcium Homeostasis, and Genetic Disorders of Calcium Metabolism. , 2010, , 1136-1159. | | 6 |
| 165 | Multiple Endocrine Neoplasia Type 1. , 2010, , 2719-2741. | | 17 |
| 166 | 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 |
| 167 | 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 |
| 168 | <i>OCRL1</i> Mutations in Dent 2 Patients Suggest a Mechanism for Phenotypic Variability. Nephron Physiology, 2009, 112, p27-p36. | 1.5 | 79 |
| 169 | Mutational Analysis of CLC-5, Cofilin and CLC-4 in Patients with Dentâ€™s Disease. Nephron Physiology, 2009, 112, p53-p62. | 1.5 | 30 |
| 170 | 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 |
| 171 | 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 |
| 172 | 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 |
| 173 | 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 |
| 174 | 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 |
| 175 | Rickets and osteomalacia. Medicine, 2009, 37, 483-488. | 0.2 | 9 |
| 176 | Multiple endocrine neoplasia. Medicine, 2009, 37, 450-453. | 0.2 | 1 |
| 177 | Genetic causes of hypercalciuric nephrolithiasis. Pediatric Nephrology, 2009, 24, 2321-2332. | 0.9 | 97 |
| 178 | Genetic Disorders of Calcium and Phosphate Homeostasis. , 2009, , 267-305. | | 1 |
| 179 | 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 |
| 180 | Gastroenteropancreatic neuroendocrine tumours. Lancet Oncology, The, 2008, 9, 61-72. | 5.1 | 1,474 |

| # | Article | IF | Citations |
|---|---|---|---|
| 181 | 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 |
| 182 | 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 |
| 183 | Familial isolated primary hyperparathyroidism caused by mutations of the MEN1 gene. Nature Clinical Practice Endocrinology and Metabolism, 2008, 4, 53-58. | 2.9 | 72 |
| 184 | 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 |
| 185 | Anatomic and Functional Imaging of Metastatic Carcinoid Tumors. Radiographics, 2007, 27, 455-477. | 1.4 | 100 |
| 186 | Cox-2 Promotes Chromogranin A Expression and Bioactivity: Evidence for a Prostaglandin E2-Dependent Mechanism and the Involvement of a Proximal Cyclic Adenosine 5â€²-Monophosphate-Responsive Element. Endocrinology, 2007, 148, 4310-4317. | 1.4 | 6 |
| 187 | A novel MEN1 intronic mutation associated with multiple endocrine neoplasia type 1. Clinical Endocrinology, 2007, 66, 709-713. | 1.2 | 12 |
| 188 | Genetics of Hypercalciuric Nephrolithiasis. Annals of the New York Academy of Sciences, 2007, 1116, 461-484. | 1.8 | 55 |
| 189 | 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 |
| 190 | Multiple Endocrine Neoplasia: Spectrum of Radiologic Appearances and Discussion of a Multitechnique Imaging Approach. Radiographics, 2006, 26, 433-451. | 1.4 | 67 |
| 191 | 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 |
| 192 | Characteristics of Hearing Loss in HDR (Hypoparathyroidism, Sensorineural Deafness, Renal Dysplasia) Syndrome. Audiology and Neuro-Otology, 2006, 11, 373-379. | 0.6 | 26 |
| 193 | Multiple Endocrine Neoplasia Type 1. , 2006, , 386-392. | | 3 |
| 194 | Dent's disease. Nephrology Dialysis Transplantation, 2005, 20, 2284-2285. | 0.4 | 5 |
| 195 | A Novel EXT1 Splice Site Mutation in a Kindred with Hereditary Multiple Exostosis and Osteoporosis. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 5386-5392. | 1.8 | 20 |
| 196 | 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 |
| 197 | MMP13 mutation causes spondyloepimetaphyseal dysplasia, Missouri type (SEMDMO). Journal of Clinical Investigation, 2005, 115, 2832-2842. | 3.9 | 89 |
| 198 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 199 | 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 |
| 200 | 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 |
| 201 | Characterization of GATA3 Mutations in the Hypoparathyroidism, Deafness, and Renal Dysplasia (HDR) Syndrome. Journal of Biological Chemistry, 2004, 279, 22624-22634. | 1.6 | 145 |
| 202 | Comparative ontogeny, processing, and segmental distribution of the renal chloride channel, ClC-5. Kidney International, 2004, 65, 198-208. | 2.6 | 27 |
| 203 | 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 |
| 204 | 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, 1060-1070. | 1.3 | 18 |
| 205 | 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 |
| 206 | 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 |
| 207 | Modeling study of human renal chloride channel (hCLC-5) mutations suggests a structural-functional relationship. Kidney International, 2003, 63, 1426-1432. | 2.6 | 49 |
| 208 | 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 |
| 209 | Genetic Mapping Studies of Familial Juvenile Hyperuricemic Nephropathy on Chromosome 16p11-p13. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 464-470. | 1.8 | 26 |
| 210 | Parathyroid Disorders. , 2003, , 485-508. | | 5 |
| 211 | Genetic contribution to renal function and electrolyte balance: a twin study. Clinical Science, 2002, 103, 259-265. | 1.8 | 104 |
| 212 | 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 |
| 213 | 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 |
| 214 | Fragmentation of filtered proteins and implications for glomerular protein sieving in Fanconi syndrome. Kidney International, 2002, 62, 349. | 2.6 | 4 |
| 215 | 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 | 27 |
| 216 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 217 | Menin Interacts Directly with the Homeobox-Containing Protein Pem. Biochemical and Biophysical Research Communications, 2001, 286, 426-431. | 1.0 | 92 |
| 218 | 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 |
| 219 | Absence of mutations in the growth hormone (GH)-releasing hormone receptor gene in GH-secreting pituitary adenomas. Clinical Endocrinology, 2001, 54, 301-307. | 1.2 | 26 |
| 220 | Glomerular protein sieving and implications for renal failure in Fanconi syndrome. Kidney International, 2001, 60, 1885-1892. | 2.6 | 207 |
| 221 | 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 |
| 222 | Mutational Analysis in X-Linked Spondyloepiphyseal Dysplasia Tarda1. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 3233-3236. | 1.8 | 21 |
| 223 | X-Linked Hypophosphatemia Attributable to Pseudoexons of the PHEX Gene. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 3840-3844. | 1.8 | 47 |
| 224 | 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 |
| 225 | X-Linked Hypophosphatemia Attributable to Pseudoexons of the PHEX Gene. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 3840-3844. | 1.8 | 10 |
| 226 | Rickets and Osteomalacia. Medicine, 2001, 29, 74-80. | 0.2 | 0 |
| 227 | Clinical and genetic studies of CLCN5 mutations in Japanese families with Dent's disease. Kidney International, 2000, 58, 520-527. | 2.6 | 54 |
| 228 | Isolated hypercalciuria with mutation in CLCN5: Relevance to idiopathic hypercalciuria. Kidney International, 2000, 57, 232-239. | 2.6 | 65 |
| 229 | Tubular proteinuria defined by a study of Dent's (CLCN5 mutation) and other tubular diseases. Kidney International, 2000, 57, 240-249. | 2.6 | 99 |
| 230 | Pathogenesis of Dent's disease and related syndromes of X-linked nephrolithiasis. Kidney International, 2000, 57, 787-793. | 2.6 | 92 |
| 231 | GATA3 haplo-insufficiency causes human HDR syndrome. Nature, 2000, 406, 419-422. | 13.7 | 516 |
| 232 | 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, 3343-3347. | 1.8 | 17 |
| 233 | 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 |
| 234 | MULTIPLE ENDOCRINE NEOPLASIA TYPE 1. Endocrinology and Metabolism Clinics of North America, 2000, 29, 541-567. | 1.2 | 80 |

| # | Article | IF | Citations |
|---|---|---|---|
| 235 | X-Linked Nephrolithiasis/Dentâ€™s Disease and Mutations in the ClC-5 Chloride Channel. , 2000, , 133-152. | | 3 |
| 236 | 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 |
| 237 | Renal Chloride Channel, CLCN5, Mutations in Dent's Disease. Journal of Bone and Mineral Research, 1999, 14, 1536-1542. | 3.1 | 32 |
| 238 | A familial syndrome due to Arg648Stop mutation in the X-linked renal chloride channel gene. Pediatric Nephrology, 1999, 13, 278-283. | 0.9 | 16 |
| 239 | Localization of Familial Benign Hypercalcemia, Oklahoma Variant (FBHOk), to Chromosome 19q13. American Journal of Human Genetics, 1999, 64, 189-195. | 2.6 | 129 |
| 240 | Genetic Disorders of Renal Electrolyte Transport. New England Journal of Medicine, 1999, 340, 1177-1187. | 13.9 | 224 |
| 241 | Clinical features of X-linked nephrolithiasis in childhood. Pediatric Nephrology, 1998, 12, 625-629. | 0.9 | 36 |
| 242 | Chloride channel mutations in hypercalciuric kidney stone disease. Clinical and Experimental Nephrology, 1998, 2, 194-198. | 0.7 | 0 |
| 243 | Expression and chromosomal localization of the Requiem gene. Mammalian Genome, 1998, 9, 660-665. | 1.0 | 22 |
| 244 | Functional characterization of renal chloride channel, CLCN5, mutations associated with Dentâ€™sJapan disease. Kidney International, 1998, 54, 1850-1856. | 2.6 | 71 |
| 245 | Mutational Analysis of PHEX Gene in X-Linked Hypophosphatemia1. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 3615-3623. | 1.8 | 104 |
| 246 | Multiple Endocrine Neoplasiaâ€”Syndromes of the Twentieth Century. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 2617-2620. | 1.8 | 131 |
| 247 | Metabolic Bone Disease in Children. , 1998, , 759-783. | | 2 |
| 248 | Characterisation of renal chloride channel, CLCN5, mutations in hypercalciuric nephrolithiasis (kidney stones) disorders. Human Molecular Genetics, 1997, 6, 1233-1239. | 1.4 | 148 |
| 249 | Expression and Cloning of the Human X-Linked Hypophosphatemia Gene cDNA. Biochemical and Biophysical Research Communications, 1997, 231, 635-639. | 1.0 | 62 |
| 250 | 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 |
| 251 | Sequence Analysis of 139 kb in Xp22.1 Containing Spermine Synthase and the 5â€² Region of PEX. Genomics, 1997, 44, 227-231. | 1.3 | 20 |
| 252 | Chloride channels cough up. Nature Genetics, 1997, 17, 125-127. | 9.4 | 28 |

| # | Article | IF | Citations |
|---|---|---|---|
| 253 | Mutations of CLCN5 in Japanese children with idiopathic low molecular weight proteinuria, hypercalciuria and nephrocalcinosis. Kidney International, 1997, 52, 911-916. | 2.6 | 82 |
| 254 | Linkage Studies of a Missouri Kindred with Autosomal Dominant Spondyloepimetaphyseal Dysplasia (SEMD) Indicate Genetic Heterogeneity. Journal of Bone and Mineral Research, 1997, 12, 1204-1209. | 3.1 | 10 |
| 255 | Definition of the MinimalMEN1Candidate Area Based on a 5-Mb Integrated Map of Proximal 11q13. Genomics, 1996, 37, 354-365. | 1.3 | 76 |
| 256 | EagI andNotI linking clones from human chromosomes 11 and Xp. Human Genetics, 1996, 97, 742-749. | 1.8 | 7 |
| 257 | Construction of a YAC contig and an STS map spanning 3.6 megabase pairs in Xp22.1. Human Genetics, 1996, 97, 60-8. | 1.8 | 16 |
| 258 | Genetic Disorders of Calcium and Phosphate Homeostasis. , 1996, , 311-345. | | 6 |
| 259 | Molecular genetics of parathyroid disease. Current Opinion in Endocrinology, Diabetes and Obesity, 1996, 3, 521-528. | 0.6 | 9 |
| 260 | A common molecular basis for three inherited kidney stone diseases. Nature, 1996, 379, 445-449. | 13.7 | 694 |
| 261 | 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 |
| 262 | 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 |
| 263 | 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 |
| 264 | Molecular genetics of disorders of calcium homeostasis. Bailliere's Clinical Endocrinology and Metabolism, 1995, 9, 581-608. | 1.0 | 9 |
| 265 | The Role of Molecular Genetics in Screening for Multiple Endocrine Neoplasia Type 1. Endocrinology and Metabolism Clinics of North America, 1994, 23, 117-135. | 1.2 | 12 |
| 266 | 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 |
| 267 | 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 |
| 268 | Autosomal Dominant Familial Hypoparathyroidism, Sensorineural Deafness, and Renal Dysplasia. New England Journal of Medicine, 1992, 327, 1069-1074. | 13.9 | 161 |
| 269 | 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 |
| 270 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 271 | Chapter 101. Genetic Basis of Renal Stones. , 0, , 474-478. | | 0 |