Caroline M Gorvin

List of Publications by Year in Descending Order

166 papers	10,608 citations	46 h-index	101 g-index
177 ext. papers	12,257 ext. citations	9.1 avg, IF	6.37 L-index

#	Paper	IF	Citations
166	Autosomal Dominant Hypocalcemia Type 1 (ADH1) Associated With Myoclonus and Intracerebral Calcifications.. *Journal of the Endocrine Society*, **2022**, 6, bvac042	0.4	0
165	The Importance of Functionally Characterizing Calcium-Sensing Receptor Variants in Individuals With Hypercalcemia.. *Journal of the Endocrine Society*, **2022**, 6, bvac052	0.4	
164	Recent advances in calcium-sensing receptor structures and signaling pathways. *Progress in Molecular Biology and Translational Science*, **2022**,	4	
163	The emerging role of heterodimerisation and interacting proteins in ghrelin receptor function. *Journal of Endocrinology*, **2021**, 252, R23-R39	4.7	1
162	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
161	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
160	Hormonal regulation of biomineralization. *Nature Reviews Endocrinology*, **2021**, 17, 261-275	15.2	14
159	CTNNB1-Mutant Aldosterone-Producing Adenomas With Somatic Mutations of GNA11/GNAQ Have Distinct Phenotype and Genotype. *Journal of the Endocrine Society*, **2021**, 5, A65-A66	0.4	78
158	The role of biomineralization in disorders of skeletal development and tooth formation. *Nature Reviews Endocrinology*, **2021**, 17, 336-349	15.2	8
157	Genetic causes of neonatal and infantile hypercalcaemia. *Pediatric Nephrology*, **2021**, 1	3.2	2
156	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
155	Multiple endocrine neoplasia type 1 in children and adolescents: Clinical features and treatment outcomes. *Surgery*, **2021**,	3.6	2
154	Asymmetric activation of the calcium-sensing receptor homodimer. *Nature*, **2021**, 595, 455-459	50.4	14
153	Calcium-sensing receptor signaling - How human disease informs biology. *Current Opinion in Endocrine and Metabolic Research*, **2021**, 16, 10-28	1.7	1
152	Small molecules restore the function of mutant CLC5 associated with Dent disease. *Journal of Cellular and Molecular Medicine*, **2021**, 25, 1319-1322	5.6	
151	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
150	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

149	Heterogeneity of G protein activation by the calcium-sensing receptor. *Journal of Molecular Endocrinology*, **2021**, 67, 41-53	4.5	3
148	The bromodomain inhibitor JQ1+ reduces calcium-sensing receptor activity in pituitary cell lines. *Journal of Molecular Endocrinology*, **2021**, 67, 83-94	4.5	
147	GPCRQ and Endocrinology **2021**,		0
146	International Union of Basic and Clinical Pharmacology. CVIII. Calcium-Sensing Receptor Nomenclature, Pharmacology, and Function. *Pharmacological Reviews*, **2020**, 72, 558-604	22.5	22
145	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
144	Genetics of kidney stone disease. *Nature Reviews Urology*, **2020**, 17, 407-421	5.5	18
143	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
142	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
141	Genetics of Skeletal Disorders. *Handbook of Experimental Pharmacology*, **2020**, 262, 325-351	3.2	0
140	Control of PTH secretion by the TRPC1 ion channel. *JCI Insight*, **2020**, 5,	9.9	2
139	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
138	Case report: a 10-year-old girl with primary hypoparathyroidism and systemic lupus erythematosus. *Journal of Pediatric Endocrinology and Metabolism*, **2020**, 33, 1231-1235	1.6	0
137	Studies of mice deleted for Sox3 and uc482: relevance to X-linked hypoparathyroidism. *Endocrine Connections*, **2020**,	3.5	3
136	ENDOCRINOLOGY IN THE TIME OF COVID-19: Clinical management of neuroendocrine neoplasms (NENs). *European Journal of Endocrinology*, **2020**, 183, G79-G88	6.5	9
135	Preclinical drug studies in MEN1-related neuroendocrine neoplasms (MEN1-NENs). *Endocrine-Related Cancer*, **2020**, 27, R345-R355	5.7	4
134	Genetic background influences tumour development in heterozygous Men1 knockout mice. *Endocrine Connections*, **2020**, 9, 426-437	3.5	2
133	Genetic regulation of parathyroid gland development **2020**, 1355-1377		
132	Activating Mutations of the G-protein Subunit 1 Interdomain Interface Cause Autosomal Dominant Hypocalcemia Type 2. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	3

131	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
130	Calcilytic NPSP795 Increases Plasma Calcium and PTH in an Autosomal Dominant Hypocalcemia Type 1 Mouse Model. *JBMR Plus*, **2020**, 4, e10402	3.9	1
129	Clinical MEN-1 Among a Large Cohort of Patients With Acromegaly. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	10
128	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
127	Mice with a Brd4 Mutation Represent a New Model of Nephrocalcinosis. *Journal of Bone and Mineral Research*, **2019**, 34, 1324-1335	6.3	3
126	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
125	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
124	Regulation of sclerostin in glucocorticoid-induced osteoporosis (GIO) in mice and humans. *Endocrine Connections*, **2019**, 8, 923-934	3.5	10
123	Molecular and clinical insights from studies of calcium-sensing receptor mutations. *Journal of Molecular Endocrinology*, **2019**, 63, R1-R16	4.5	19
122	Genetic variants of calcium and vitamin D metabolism in kidney stone disease. *Nature Communications*, **2019**, 10, 5175	17.4	27
121	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
120	Association of prolactin receptor (PRLR) variants with prolactinomas. *Human Molecular Genetics*, **2019**, 28, 1023-1037	5.6	12
119	Genetic approaches to metabolic bone diseases. *British Journal of Clinical Pharmacology*, **2019**, 85, 1147-1160	3.8	11
118	A calcium-sensing receptor mutation causing hypocalcemia disrupts a transmembrane salt bridge to activate β-arrestin-biased signaling. *Science Signaling*, **2018**, 11,	8.8	26
117	Vascular wall regulator of G-protein signalling-1 (RGS-1) is required for angiotensin II-mediated blood pressure control. *Vascular Pharmacology*, **2018**, 108, 15-22	5.9	7
116	Current and emerging therapies for PNETs in patients with or without MEN1. *Nature Reviews Endocrinology*, **2018**, 14, 216-227	15.2	34
115	AP2σ 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
114	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

113	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
112	Jeffrey Lima Hayes O□iordan: March 27, 1931 □October 9, 2017. *Journal of Bone and Mineral Research*, **2018**, 33, 1-2	6.3	3
111	Insights into calcium-sensing receptor trafficking and biased signalling by studies of calcium homeostasis. *Journal of Molecular Endocrinology*, **2018**, 61, R1-R12	4.5	14
110	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
109	Introduction to Genetics of Skeletal and Mineral Metabolic Diseases **2018**, 1-21		2
108	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
107	Molecular Genetic Studies of Pancreatic Neuroendocrine Tumors: New Therapeutic Approaches. *Endocrinology and Metabolism Clinics of North America*, **2018**, 47, 525-548	5.5	14
106	Further delineation of Malan syndrome. *Human Mutation*, **2018**, 39, 1226-1237	4.7	24
105	MiR-15a/miR-16-1 expression inversely correlates with cyclin D1 levels in Men1 pituitary NETs. *Journal of Endocrinology*, **2018**,	4.7	9
104	The calcium-sensing receptor in physiology and in calcitropic and noncalcitropic diseases. *Nature Reviews Endocrinology*, **2018**, 15, 33-51	15.2	113
103	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
102	Introduction to Genetics **2018**, 341-350		
101	Familial States of Primary Hyperparathyroidism **2018**, 629-638		0
100	Hypoparathyroidism **2018**, 617-636		
99	Expression profiling of colorectal cancer cells reveals inhibition of DNA replication licensing by extracellular calcium. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2017**, 1864, 987-996	4.9	7
98	A mouse model for inherited renal fibrosis associated with endoplasmic reticulum stress. *DMM Disease Models and Mechanisms*, **2017**, 10, 773-786	4.1	26
97	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
96	Knockin mouse with mutant G mimics human inherited hypocalcemia and is rescued by pharmacologic inhibitors. *JCI Insight*, **2017**, 2, e91079	9.9	23

95	Hypoparathyroidism. *Nature Reviews Disease Primers*, **2017**, 3, 17055	51.1	100
94	Molecular genetics of syndromic and non-syndromic forms of parathyroid carcinoma. *Human Mutation*, **2017**, 38, 1621-1648	4.7	55
93	Hypercalcemic Disorders in Children. *Journal of Bone and Mineral Research*, **2017**, 32, 2157-2170	6.3	51
92	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
91	Mutant Mice With Calcium-Sensing Receptor Activation Have Hyperglycemia That Is Rectified by Calcilytic Therapy. *Endocrinology*, **2017**, 158, 2486-2502	4.8	21
90	G mutation in mice causes hypocalcemia rectifiable by calcilytic therapy. *JCI Insight*, **2017**, 2, e91103	9.9	25
89	Genomic profiling reveals mutational landscape in parathyroid carcinomas. *JCI Insight*, **2017**, 2, e92061	9.9	50
88	Cinacalcet corrects hypercalcemia in mice with an inactivating G□1 mutation. *JCI Insight*, **2017**, 2,	9.9	10
87	Regulation of Calcium Homeostasis and Genetic Disorders that Affect Calcium Metabolism **2016**, 1063-1089.e10		4
86	Autosomal dominant osteopetrosis associated with renal tubular acidosis is due to a CLCN7 mutation. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2988-2992	2.5	5
85	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
84	Cinacalcet for Symptomatic Hypercalcemia Caused by AP2S1 Mutations. *New England Journal of Medicine*, **2016**, 374, 1396-1398	59.2	29
83	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
82	Management of Hypoparathyroidism: Summary Statement and Guidelines. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 2273-83	5.6	207
81	Presentation of Hypoparathyroidism: Etiologies and Clinical Features. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 2300-12	5.6	175
80	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
79	Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis **2016**, 291-339		
78	A G-protein Subunit-□1 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

77	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
76	Novel gene function revealed by mouse mutagenesis screens for models of age-related disease. *Nature Communications*, **2016**, 7, 12444	17.4	56
75	Pseudohypoparathyroidism type 1a due to a novel mutation in the GNAS gene. *Clinical Endocrinology*, **2016**, 84, 463-5	3.4	4
74	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
73	Identification of a G-Protein Subunit-11 Gain-of-Function Mutation, Val340Met, in a Family With Autosomal Dominant Hypocalcemia Type 2 (ADH2). *Journal of Bone and Mineral Research*, **2016**, 31, 1207-14	6.3	33
72	The calcium-sensing receptor: And its involvement in parathyroid pathology. *Annales DœEndocrinologie*, **2015**, 76, 81-3	1.7	9
71	The prolactin receptor: Diverse and emerging roles in pathophysiology. *Journal of Clinical and Translational Endocrinology*, **2015**, 2, 85-91	2.4	21
70	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
69	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. *Nature Genetics*, **2015**, 47, 717-726	36.3	244
68	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
67	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
66	Familial and Hereditary Forms of Primary Hyperparathyroidism **2015**, 341-363		6
65	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
64	SaO021A NOVEL ROLE FOR GATA3 IN MESANGIAL CELLS IN GLOMERULAR DEVELOPMENT AND INJURY. *Nephrology Dialysis Transplantation*, **2015**, 30, iii32-iii32	4.3	1
63	Molecular genetic advances in pituitary tumor development. *Expert Review of Endocrinology and Metabolism*, **2015**, 10, 35-53	4.1	5
62	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
61	Multiple endocrine neoplasia type 1 (MEN1) and type 4 (MEN4). *Molecular and Cellular Endocrinology*, **2014**, 386, 2-15	4.4	261
60	Mutant prolactin receptor and familial hyperprolactinemia. *New England Journal of Medicine*, **2014**, 370, 977-8	59.2	9

59	Diagnosis of asymptomatic primary hyperparathyroidism: proceedings of the Fourth International Workshop. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, 3570-9	5.6	233
58	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
57	Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis **2014**, 1-59		
56	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
55	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
54	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
53	Mutant prolactin receptor and familial hyperprolactinemia. *New England Journal of Medicine*, **2013**, 369, 2012-2020	59.2	80
52	Investigating hypocalcaemia. *BMJ, The*, **2013**, 346, f2213	5.9	34
51	Mutations affecting G-protein subunit α11 in hypercalcemia and hypocalcemia. *New England Journal of Medicine*, **2013**, 368, 2476-2486	59.2	282
50	Whole-exome sequencing studies of nonfunctioning pituitary adenomas. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, E796-800	5.6	69
49	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
48	Mutations in AP2S1 cause familial hypocalciuric hypercalcemia type 3. *Nature Genetics*, **2013**, 45, 93-7	36.3	196
47	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
46	A mouse model of early-onset renal failure due to a xanthine dehydrogenase nonsense mutation. *PLoS ONE*, **2012**, 7, e45217	3.7	6
45	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
44	Clinical practice guidelines for multiple endocrine neoplasia type 1 (MEN1). *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, 2990-3011	5.6	830
43	Whole-exome sequencing studies of nonhereditary (sporadic) parathyroid adenomas. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E1995-2005	5.6	101
42	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

41	Proliferation rates of multiple endocrine neoplasia type 1 (MEN1)-associated tumors. *Endocrinology* , **2012**, 153, 5167-79	4.8	13
40	Bone Mineral Content and Density. *Current Protocols in Mouse Biology*, **2012**, 2, 365-400	1.1	5
39	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
38	Mouse models for inherited endocrine and metabolic disorders. *Journal of Endocrinology*, **2011**, 211, 211-30	4.7	28
37	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
36	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
35	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
34	CLC-5 and KIF3B interact to facilitate CLC-5 plasma membrane expression, endocytosis, and microtubular transport: relevance to pathophysiology of Dent disease. *American Journal of Physiology - Renal Physiology*, **2010**, 298, F365-80	4.3	52
33	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
32	Dent disease. *Orphanet Journal of Rare Diseases*, **2010**, 5, 28	4.2	133
31	Multiple endocrine neoplasia type 1 (MEN1). *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2010**, 24, 355-70	6.5	161
30	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
29	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
28	Mutational analysis of CLC-5, cofilin and CLC-4 in patients with Dent disease. *Nephron Physiology*, **2009**, 112, p53-62		26
27	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
26	Characterization of Dent 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
25	Genetic causes of hypercalciuric nephrolithiasis. *Pediatric Nephrology*, **2009**, 24, 2321-32	3.2	79
24	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

23	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
22	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
21	The ClC-5 knockout mouse model of Dent disease has renal hypercalciuria and increased bone turnover. *Journal of Bone and Mineral Research*, **2003**, 18, 615-23	6.3	65
20	Genetic contribution to renal function and electrolyte balance: a twin study. *Clinical Science*, **2002**, 103, 259-65	6.5	86
19	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
18	GATA3 haplo-insufficiency causes human HDR syndrome. *Nature*, **2000**, 406, 419-22	50.4	452
17	Mice lacking renal chloride channel, CLC-5, are a model for Dent disease, a nephrolithiasis disorder associated with defective receptor-mediated endocytosis. *Human Molecular Genetics*, **2000** , 9, 2937-45	5.6	248
16	Characterization of renal chloride channel (CLCN5) mutations in Dent disease. *Journal of the American Society of Nephrology: JASN*, **2000**, 11, 1460-1468	12.7	40
15	Renal chloride channel, CLCN5, mutations in Dent disease. *Journal of Bone and Mineral Research*, **1999**, 14, 1536-42	6.3	27
14	A familial syndrome due to Arg648Stop mutation in the X-linked renal chloride channel gene. *Pediatric Nephrology*, **1999**, 13, 278-83	3.2	14
13	Localization of familial benign hypercalcemia, Oklahoma variant (FBHOk), to chromosome 19q13. *American Journal of Human Genetics*, **1999**, 64, 189-95	11	119
12	Clinical features of X-linked nephrolithiasis in childhood. *Pediatric Nephrology*, **1998**, 12, 625-9	3.2	27
11	Chloride channel mutations in hypercalciuric kidney stone disease. *Clinical and Experimental Nephrology*, **1998**, 2, 194-198	2.5	
10	Expression and chromosomal localization of the Requiem gene. *Mammalian Genome*, **1998**, 9, 660-5	3.2	18
9	Characterisation of renal chloride channel, CLCN5, mutations in hypercalciuric nephrolithiasis (kidney stones) disorders. *Human Molecular Genetics*, **1997**, 6, 1233-9	5.6	125
8	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
7	EagI and NotI linking clones from human chromosomes 11 and Xp. *Human Genetics*, **1996**, 97, 742-9	6.3	7
6	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

5	A common molecular basis for three inherited kidney stone diseases. *Nature*, **1996**, 379, 445-9	50.4	614
4	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
3	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
2	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
1	Frequent Occurrence of an Intron 4 Mutation in Multiple Endocrine Neoplasia Type 1		11