Caroline M Gorvin

List of Publications by Year
in descending order

169 papers	14,232 citations	38660 50 h-index	22102 113 g-index
179 all docs	179 docs citations	179 times ranked	10644 citing authors

#	Article	IF	Citations
1	Recent advances in calcium-sensing receptor structures and signaling pathways. Progress in Molecular Biology and Translational Science, 2023, , 121-135.	0.9	2
2	Genetic causes of neonatal and infantile hypercalcaemia. Pediatric Nephrology, 2022, 37, 289-301.	0.9	10
3	The emerging role of heterodimerisation and interacting proteins in ghrelin receptor function. Journal of Endocrinology, 2022, 252, R23-R39.	1.2	8
4	Autosomal Dominant Hypocalcemia Type 1 (ADH1) Associated With Myoclonus and Intracerebral Calcifications. Journal of the Endocrine Society, 2022, 6, bvac042.	0.1	5
5	The importance of functionally characterizing calcium-sensing receptor variants in individuals with hypercalcemia. Journal of the Endocrine Society, 2022, 6, bvac052.	0.1	0
6	Genetics of monogenic disorders of calcium and bone metabolism. Clinical Endocrinology, 2022, 97, 483-501.	1.2	7
7	Spectrum of germline AIRE mutations causing APS-1 and familial hypoparathyroidism. European Journal of Endocrinology, 2022, , .	1.9	1
8	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
9	Skeletal and extraskeletal disorders of biomineralization. Nature Reviews Endocrinology, 2022, 18, 473-489.	4.3	25
10	miR-3156-5p is downregulated in serum of MEN1 patients and regulates expression of MORF4L2. Endocrine-Related Cancer, 2022, 29, 557-568.	1.6	5
11	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
12	Calcium-sensing receptor signaling â€” How human disease informs biology. Current Opinion in Endocrine and Metabolic Research, 2021, 16, 10-18.	0.6	7
13	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
14	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
15	Hormonal regulation of biomineralization. Nature Reviews Endocrinology, 2021, 17, 261-275.	4.3	50
16	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.	0.1	0
17	The role of biomineralization in disorders of skeletal development and tooth formation. Nature Reviews Endocrinology, 2021, 17, 336-349.	4.3	46
18	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

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19	Multiple endocrine neoplasia type 1 in children and adolescents: Clinical features and treatment outcomes. Surgery, 2021, , .	1.0	10
20	Asymmetric activation of the calcium-sensing receptor homodimer. Nature, 2021, 595, 455-459.	13.7	59
21	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
22	Heterogeneity of G protein activation by the calcium-sensing receptor. Journal of Molecular Endocrinology, 2021, 67, 41-53.	1.1	4
23	The bromodomain inhibitor JQ1+ reduces calcium-sensing receptor activity in pituitary cell lines. Journal of Molecular Endocrinology, 2021, 67, 83-94.	1.1	1
24	GPCR's and Endocrinology. , 2021, , .		1
25	Genetic regulation of parathyroid gland development. , 2020, , 1355-1377.		0
26	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
27	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
28	Calcilytic NPSP795 Increases Plasma Calcium and PTH in an Autosomal Dominant Hypocalcemia Type 1 Mouse Model. JBMR Plus, 2020, 4, e10402.	1.3	3
29	International Union of Basic and Clinical Pharmacology. CVIII. Calcium-Sensing Receptor Nomenclature, Pharmacology, and Function. Pharmacological Reviews, 2020, 72, 558-604.	7.1	59
30	Hypoparathyroidism, deafness, and renal dysplasia syndrome: 20 Years after the identification of the firstGATA3mutations. Human Mutation, 2020, 41, 1341-1350.	1.1	19
31	Genetics of kidney stone disease. Nature Reviews Urology, 2020, 17, 407-421.	1.9	81
32	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
33	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
34	Genetics of Skeletal Disorders. Handbook of Experimental Pharmacology, 2020, 262, 325-351.	0.9	3
35	Clinical MEN-1 Among a Large Cohort of Patients With Acromegaly. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e2271-e2281.	1.8	19
36	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

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37	Control of PTH secretion by the TRPC1 ion channel. JCI Insight, 2020, 5, .	2.3	6
38	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
39	Case report: a 10-year-old girl with primary hypoparathyroidism and systemic lupus erythematosus. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1231-1235.	0.4	2
40	Studies of mice deleted for Sox3 and uc482: relevance to X-linked hypoparathyroidism. Endocrine Connections, 2020, 9, 173-186.	0.8	3
41	ENDOCRINOLOGY IN THE TIME OF COVID-19: Clinical management of neuroendocrine neoplasms (NENs). European Journal of Endocrinology, 2020, 183, G79-G88.	1.9	11
42	Preclinical drug studies in MEN1-related neuroendocrine neoplasms (MEN1-NENs). Endocrine-Related Cancer, 2020, 27, R345-R355.	1.6	5
43	Genetic background influences tumour development in heterozygous Men1 knockout mice. Endocrine Connections, 2020, 9, 426-437.	0.8	5
44	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
45	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
46	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
47	Mice with a Brd4 Mutation Represent a New Model of Nephrocalcinosis. Journal of Bone and Mineral Research, 2019, 34, 1324-1335.	3.1	7
48	Genetic variants of calcium and vitamin D metabolism in kidney stone disease. Nature Communications, 2019, 10, 5175.	5.8	69
49	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
50	Association of prolactin receptor (PRLR) variants with prolactinomas. Human Molecular Genetics, 2019, 28, 1023-1037.	1.4	24
51	The calcium-sensing receptor in physiology and in calcitropic and noncalcitropic diseases. Nature Reviews Endocrinology, 2019, 15, 33-51.	4.3	226
52	Genetic approaches to metabolic bone diseases. British Journal of Clinical Pharmacology, 2019, 85, 1147-1160.	1.1	21
53	Regulation of sclerostin in glucocorticoid-induced osteoporosis (GIO) in mice and humans. Endocrine Connections, 2019, 8, 923-934.	0.8	15
54	Molecular and clinical insights from studies of calcium-sensing receptor mutations. Journal of Molecular Endocrinology, 2019, 63, R1-R16.	1.1	35

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55	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
56	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
57	Vascular wall regulator of G-protein signalling-1 (RGS-1) is required for angiotensin IIâ€“mediated blood pressure control. Vascular Pharmacology, 2018, 108, 15-22.	1.0	13
58	Current and emerging therapies for PNETs in patients with or without MEN1. Nature Reviews Endocrinology, 2018, 14, 216-227.	4.3	46
59	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
60	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
61	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
62	Jeffrey Lima Hayes O'Riordan: March 27, 1931 â€“ October 9, 2017. Journal of Bone and Mineral Research, 2018, 33, 1-2.	3.1	25
63	Insights into calcium-sensing receptor trafficking and biased signalling by studies of calcium homeostasis. Journal of Molecular Endocrinology, 2018, 61, R1-R12.	1.1	19
64	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
65	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
66	Hypoparathyroidism. , 2018, , 617-636.		0
67	Introduction to Genetics of Skeletal and Mineral Metabolic Diseases. , 2018, , 1-21.		2
68	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
69	Molecular Genetic Studies of Pancreatic Neuroendocrine Tumors. Endocrinology and Metabolism Clinics of North America, 2018, 47, 525-548.	1.2	17
70	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	1.1	42
71	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.	1.9	8
72	Mouse model for inherited renal fibrosis associated with endoplasmic reticulum stress. DMM Disease Models and Mechanisms, 2017, 10, 773-786.	1.2	34

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73	Hypoparathyroidism. Nature Reviews Disease Primers, 2017, 3, 17055.	18.1	142
74	Molecular genetics of syndromic and non-syndromic forms of parathyroid carcinoma. Human Mutation, 2017, 38, 1621-1648.	1.1	82
75	Hypercalcemic Disorders in Children. Journal of Bone and Mineral Research, 2017, 32, 2157-2170.	3.1	82
76	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
77	Mutant Mice With Calcium-Sensing Receptor Activation Have Hyperglycemia That Is Rectified by Calcilytic Therapy. Endocrinology, 2017, 158, 2486-2502.	1.4	31
78	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
79	Knockin mouse with mutant GÎ±11 mimics human inherited hypocalcemia and is rescued by pharmacologic inhibitors. JCI Insight, 2017, 2, e91079.	2.3	26
80	GÎ±11 mutation in mice causes hypocalcemia rectifiable by calcilytic therapy. JCI Insight, 2017, 2, e91103.	2.3	28
81	Genomic profiling reveals mutational landscape in parathyroid carcinomas. JCI Insight, 2017, 2, e92061.	2.3	84
82	Cinacalcet corrects hypercalcemia in mice with an inactivating GÎ±11 mutation. JCI Insight, 2017, 2, .	2.3	17
83	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
84	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
85	Novel gene function revealed by mouse mutagenesis screens for models of age-related disease. Nature Communications, 2016, 7, 12444.	5.8	79
86	Pseudohypoparathyroidism type 1a due to a novel mutation in the <scp>GNAS</scp> gene. Clinical Endocrinology, 2016, 84, 463-465.	1.2	4
87	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
88	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
89	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.	0.7	10
90	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

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91	Cinacalcet for Symptomatic Hypercalcemia Caused by AP2S1 Mutations. New England Journal of Medicine, 2016, 374, 1396-1398.	13.9	38
92	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
93	Management of Hypoparathyroidism: Summary Statement and Guidelines. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2273-2283.	1.8	303
94	Presentation of Hypoparathyroidism: Etiologies and Clinical Features. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2300-2312.	1.8	246
95	Regulation of Calcium Homeostasis and Genetic Disorders that Affect Calcium Metabolism. , 2016, , 1063-1089.e10.		7
96	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
97	Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis. , 2016, , 291-339.		0
98	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
99	SaO021A NOVEL ROLE FOR GATA3 IN MESANGIAL CELLS IN GLOMERULAR DEVELOPMENT AND INJURY. Nephrology Dialysis Transplantation, 2015, 30, iii32-iii32.	0.4	1
100	Molecular genetic advances in pituitary tumor development. Expert Review of Endocrinology and Metabolism, 2015, 10, 35-53.	1.2	5
101	The calcium-sensing receptor: And its involvement in parathyroid pathology. Annales D'Endocrinologie, 2015, 76, 81-83.	0.6	10
102	The prolactin receptor: Diverse and emerging roles in pathophysiology. Journal of Clinical and Translational Endocrinology, 2015, 2, 85-91.	1.0	31
103	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
104	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nature Genetics, 2015, 47, 717-726.	9.4	310
105	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
106	The Calcilytic Agent NPS 2143 Rectifies Hypocalcemia in a Mouse Model With an Activating Calcium-Sensing Receptor (CaSR) Mutation: Relevance to Autosomal Dominant Hypocalcemia Type 1 (ADH1). Endocrinology, 2015, 156, 3114-3121.	1.4	55
107	Familial and Hereditary Forms of Primary Hyperparathyroidism. , 2015, , 341-363.		7
108	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

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109	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
110	Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis. , 2014, , 1-59.		0
111	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
112	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
113	Multiple endocrine neoplasia type 1 (MEN1) and type 4 (MEN4). Molecular and Cellular Endocrinology, 2014, 386, 2-15.	1.6	334
114	Mutant Prolactin Receptor and Familial Hyperprolactinemia. New England Journal of Medicine, 2014, 370, 976-978.	13.9	9
115	Diagnosis of Asymptomatic Primary Hyperparathyroidism: Proceedings of the Fourth International Workshop. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 3570-3579.	1.8	296
116	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
117	Mutant Prolactin Receptor and Familial Hyperprolactinemia. New England Journal of Medicine, 2013, 369, 2012-2020.	13.9	106
118	Investigating hypocalcaemia. BMJ, The, 2013, 346, f2213-f2213.	3.0	44
119	Mutations Affecting G-Protein Subunit Î±₁₁in Hypercalcemia and Hypocalcemia. New England Journal of Medicine, 2013, 368, 2476-2486.	13.9	340
120	Whole-Exome Sequencing Studies of Nonfunctioning Pituitary Adenomas. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E796-E800.	1.8	85
121	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
122	Mutations in AP2S1 cause familial hypocalciuric hypercalcemia type 3. Nature Genetics, 2013, 45, 93-97.	9.4	242
123	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
124	Whole-Exome Sequencing Studies of Nonhereditary (Sporadic) Parathyroid Adenomas. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1995-E2005.	1.8	121
125	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
126	Proliferation Rates of Multiple Endocrine Neoplasia Type 1 (MEN1)-Associated Tumors. Endocrinology, 2012, 153, 5167-5179.	1.4	13

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127	A Mouse Model of Early-Onset Renal Failure Due to a Xanthine Dehydrogenase Nonsense Mutation. PLoS ONE, 2012, 7, e45217.	1.1	8
128	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
129	Clinical Practice Guidelines for Multiple Endocrine Neoplasia Type 1 (MEN1). Journal of Clinical Endocrinology and Metabolism, 2012, 97, 2990-3011.	1.8	1,127
130	Bone Mineral Content and Density. , 2012, 2, 365-400.		9
131	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
132	Mouse models for inherited endocrine and metabolic disorders. Journal of Endocrinology, 2011, 211, 211-230.	1.2	30
133	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
134	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
135	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
136	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
137	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
138	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
139	Dent's disease. Orphanet Journal of Rare Diseases, 2010, 5, 28.	1.2	181
140	Multiple endocrine neoplasia type 1 (MEN1). Best Practice and Research in Clinical Endocrinology and Metabolism, 2010, 24, 355-370.	2.2	206
141	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
142	Mutational Analysis of CLC-5, Cofilin and CLC-4 in Patients with Dentâ€™s Disease. Nephron Physiology, 2009, 112, p53-p62.	1.5	30
143	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
144	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

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145	Genetic causes of hypercalciuric nephrolithiasis. Pediatric Nephrology, 2009, 24, 2321-2332.	0.9	97
146	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
147	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
148	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
149	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
150	Genetic contribution to renal function and electrolyte balance: a twin study. Clinical Science, 2002, 103, 259-265.	1.8	104
151	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
152	GATA3 haplo-insufficiency causes human HDR syndrome. Nature, 2000, 406, 419-422.	13.7	516
153	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
154	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
155	Renal Chloride Channel, CLCN5, Mutations in Dent's Disease. Journal of Bone and Mineral Research, 1999, 14, 1536-1542.	3.1	32
156	A familial syndrome due to Arg648Stop mutation in the X-linked renal chloride channel gene. Pediatric Nephrology, 1999, 13, 278-283.	0.9	16
157	Localization of Familial Benign Hypercalcemia, Oklahoma Variant (FBHOk), to Chromosome 19q13. American Journal of Human Genetics, 1999, 64, 189-195.	2.6	129
158	Clinical features of X-linked nephrolithiasis in childhood. Pediatric Nephrology, 1998, 12, 625-629.	0.9	36
159	Chloride channel mutations in hypercalciuric kidney stone disease. Clinical and Experimental Nephrology, 1998, 2, 194-198.	0.7	0
160	Expression and chromosomal localization of the Requiem gene. Mammalian Genome, 1998, 9, 660-665.	1.0	22
161	Characterisation of renal chloride channel, CLCN5, mutations in hypercalciuric nephrolithiasis (kidney stones) disorders. Human Molecular Genetics, 1997, 6, 1233-1239.	1.4	148
162	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

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