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	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
2	Clinical Practice Guidelines for Multiple Endocrine Neoplasia Type 1 (MEN1). Journal of Clinical Endocrinology and Metabolism, 2012, 97, 2990-3011.	1.8	1,127
3	A common molecular basis for three inherited kidney stone diseases. Nature, 1996, 379, 445-449.	13.7	694
4	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
5	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
6	GATA3 haplo-insufficiency causes human HDR syndrome. Nature, 2000, 406, 419-422.	13.7	516
7	Mutations Affecting G-Protein Subunit Î±₁₁in Hypercalcemia and Hypocalcemia. New England Journal of Medicine, 2013, 368, 2476-2486.	13.9	340
8	Multiple endocrine neoplasia type 1 (MEN1) and type 4 (MEN4). Molecular and Cellular Endocrinology, 2014, 386, 2-15.	1.6	334
9	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nature Genetics, 2015, 47, 717-726.	9.4	310
10	Management of Hypoparathyroidism: Summary Statement and Guidelines. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2273-2283.	1.8	303
11	Diagnosis of Asymptomatic Primary Hyperparathyroidism: Proceedings of the Fourth International Workshop. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 3570-3579.	1.8	296
12	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
13	Presentation of Hypoparathyroidism: Etiologies and Clinical Features. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2300-2312.	1.8	246
14	Mutations in AP2S1 cause familial hypocalciuric hypercalcemia type 3. Nature Genetics, 2013, 45, 93-97.	9.4	242
15	The calcium-sensing receptor in physiology and in calcitropic and noncalcitropic diseases. Nature Reviews Endocrinology, 2019, 15, 33-51.	4.3	226
16	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
17	Multiple endocrine neoplasia type 1 (MEN1). Best Practice and Research in Clinical Endocrinology and Metabolism, 2010, 24, 355-370.	2.2	206
18	Dent's disease. Orphanet Journal of Rare Diseases, 2010, 5, 28.	1.2	181

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19	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
20	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
21	Characterisation of renal chloride channel, CLCN5, mutations in hypercalciuric nephrolithiasis (kidney stones) disorders. Human Molecular Genetics, 1997, 6, 1233-1239.	1.4	148
22	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
23	Hypoparathyroidism. Nature Reviews Disease Primers, 2017, 3, 17055.	18.1	142
24	Localization of Familial Benign Hypercalcemia, Oklahoma Variant (FBHOk), to Chromosome 19q13. American Journal of Human Genetics, 1999, 64, 189-195.	2.6	129
25	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
26	Whole-Exome Sequencing Studies of Nonhereditary (Sporadic) Parathyroid Adenomas. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1995-E2005.	1.8	121
27	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
28	Mutant Prolactin Receptor and Familial Hyperprolactinemia. New England Journal of Medicine, 2013, 369, 2012-2020.	13.9	106
29	Genetic contribution to renal function and electrolyte balance: a twin study. Clinical Science, 2002, 103, 259-265.	1.8	104
30	Genetic causes of hypercalciuric nephrolithiasis. Pediatric Nephrology, 2009, 24, 2321-2332.	0.9	97
31	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
32	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
33	Whole-Exome Sequencing Studies of Nonfunctioning Pituitary Adenomas. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E796-E800.	1.8	85
34	Genomic profiling reveals mutational landscape in parathyroid carcinomas. JCI Insight, 2017, 2, e92061.	2.3	84
35	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
36	Molecular genetics of syndromic and non-syndromic forms of parathyroid carcinoma. Human Mutation, 2017, 38, 1621-1648.	1.1	82

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37	Hypercalcemic Disorders in Children. Journal of Bone and Mineral Research, 2017, 32, 2157-2170.	3.1	82
38	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
39	Genetics of kidney stone disease. Nature Reviews Urology, 2020, 17, 407-421.	1.9	81
40	Novel gene function revealed by mouse mutagenesis screens for models of age-related disease. Nature Communications, 2016, 7, 12444.	5.8	79
41	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
42	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
43	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
44	Genetic variants of calcium and vitamin D metabolism in kidney stone disease. Nature Communications, 2019, 10, 5175.	5.8	69
45	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
46	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
47	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
48	International Union of Basic and Clinical Pharmacology. CVIII. Calcium-Sensing Receptor Nomenclature, Pharmacology, and Function. Pharmacological Reviews, 2020, 72, 558-604.	7.1	59
49	Asymmetric activation of the calcium-sensing receptor homodimer. Nature, 2021, 595, 455-459.	13.7	59
50	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
51	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
52	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
53	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
54	Hormonal regulation of biomineralization. Nature Reviews Endocrinology, 2021, 17, 261-275.	4.3	50

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55	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
56	Current and emerging therapies for PNETs in patients with or without MEN1. Nature Reviews Endocrinology, 2018, 14, 216-227.	4.3	46
57	The role of biomineralization in disorders of skeletal development and tooth formation. Nature Reviews Endocrinology, 2021, 17, 336-349.	4.3	46
58	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
59	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
60	Investigating hypocalcaemia. BMJ, The, 2013, 346, f2213-f2213.	3.0	44
61	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	1.1	42
62	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
63	Cinacalcet for Symptomatic Hypercalcemia Caused by AP2S1 Mutations. New England Journal of Medicine, 2016, 374, 1396-1398.	13.9	38
64	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
65	Clinical features of X-linked nephrolithiasis in childhood. Pediatric Nephrology, 1998, 12, 625-629.	0.9	36
66	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
67	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
68	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
69	Molecular and clinical insights from studies of calcium-sensing receptor mutations. Journal of Molecular Endocrinology, 2019, 63, R1-R16.	1.1	35
70	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
71	Mouse model for inherited renal fibrosis associated with endoplasmic reticulum stress. DMM Disease Models and Mechanisms, 2017, 10, 773-786.	1.2	34
72	Renal Chloride Channel, CLCN5, Mutations in Dent's Disease. Journal of Bone and Mineral Research, 1999, 14, 1536-1542.	3.1	32

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73	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
74	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
75	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
76	The prolactin receptor: Diverse and emerging roles in pathophysiology. Journal of Clinical and Translational Endocrinology, 2015, 2, 85-91.	1.0	31
77	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
78	Mutant Mice With Calcium-Sensing Receptor Activation Have Hyperglycemia That Is Rectified by Calcilytic Therapy. Endocrinology, 2017, 158, 2486-2502.	1.4	31
79	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
80	Mutational Analysis of CLC-5, Cofilin and CLC-4 in Patients with Dentâ€™s Disease. Nephron Physiology, 2009, 112, p53-p62.	1.5	30
81	Mouse models for inherited endocrine and metabolic disorders. Journal of Endocrinology, 2011, 211, 211-230.	1.2	30
82	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
83	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
84	GÎ±11 mutation in mice causes hypocalcemia rectifiable by calcilytic therapy. JCI Insight, 2017, 2, e91103.	2.3	28
85	Frequent Occurrence of an Intron 4 Mutation in Multiple Endocrine Neoplasia Type 1. , 0, .		27
86	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
87	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
88	Knockin mouse with mutant GÎ±11 mimics human inherited hypocalcemia and is rescued by pharmacologic inhibitors. JCI Insight, 2017, 2, e91079.	2.3	26
89	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
90	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

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91	Skeletal and extraskeletal disorders of biomineralization. Nature Reviews Endocrinology, 2022, 18, 473-489.	4.3	25
92	Association of prolactin receptor (PRLR) variants with prolactinomas. Human Molecular Genetics, 2019, 28, 1023-1037.	1.4	24
93	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
94	Expression and chromosomal localization of the Requiem gene. Mammalian Genome, 1998, 9, 660-665.	1.0	22
95	Genetic approaches to metabolic bone diseases. British Journal of Clinical Pharmacology, 2019, 85, 1147-1160.	1.1	21
96	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
97	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
98	Hypoparathyroidism, deafness, and renal dysplasia syndrome: 20 Years after the identification of the firstGATA3mutations. Human Mutation, 2020, 41, 1341-1350.	1.1	19
99	Clinical MEN-1 Among a Large Cohort of Patients With Acromegaly. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e2271-e2281.	1.8	19
100	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
101	Molecular Genetic Studies of Pancreatic Neuroendocrine Tumors. Endocrinology and Metabolism Clinics of North America, 2018, 47, 525-548.	1.2	17
102	Cinacalcet corrects hypercalcemia in mice with an inactivating GÎ±11 mutation. JCI Insight, 2017, 2, .	2.3	17
103	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
104	A familial syndrome due to Arg648Stop mutation in the X-linked renal chloride channel gene. Pediatric Nephrology, 1999, 13, 278-283.	0.9	16
105	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
106	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
107	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
108	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

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109	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
110	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
111	Regulation of sclerostin in glucocorticoid-induced osteoporosis (GIO) in mice and humans. Endocrine Connections, 2019, 8, 923-934.	0.8	15
112	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
113	Proliferation Rates of Multiple Endocrine Neoplasia Type 1 (MEN1)-Associated Tumors. Endocrinology, 2012, 153, 5167-5179.	1.4	13
114	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
115	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
116	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
117	ENDOCRINOLOGY IN THE TIME OF COVID-19: Clinical management of neuroendocrine neoplasms (NENs). European Journal of Endocrinology, 2020, 183, G79-G88.	1.9	11
118	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
119	The calcium-sensing receptor: And its involvement in parathyroid pathology. Annales D'Endocrinologie, 2015, 76, 81-83.	0.6	10
120	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
121	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
122	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
123	Genetic causes of neonatal and infantile hypercalcaemia. Pediatric Nephrology, 2022, 37, 289-301.	0.9	10
124	Multiple endocrine neoplasia type 1 in children and adolescents: Clinical features and treatment outcomes. Surgery, 2021, , .	1.0	10
125	Mutant Prolactin Receptor and Familial Hyperprolactinemia. New England Journal of Medicine, 2014, 370, 976-978.	13.9	9
126	Bone Mineral Content and Density. , 2012, 2, 365-400.		9

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127	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
128	A Mouse Model of Early-Onset Renal Failure Due to a Xanthine Dehydrogenase Nonsense Mutation. PLoS ONE, 2012, 7, e45217.	1.1	8
129	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
130	The emerging role of heterodimerisation and interacting proteins in ghrelin receptor function. Journal of Endocrinology, 2022, 252, R23-R39.	1.2	8
131	EagI andNotI linking clones from human chromosomes 11 and Xp. Human Genetics, 1996, 97, 742-749.	1.8	7
132	Familial and Hereditary Forms of Primary Hyperparathyroidism. , 2015, , 341-363.		7
133	Regulation of Calcium Homeostasis and Genetic Disorders that Affect Calcium Metabolism. , 2016, , 1063-1089.e10.		7
134	Mice with a Brd4 Mutation Represent a New Model of Nephrocalcinosis. Journal of Bone and Mineral Research, 2019, 34, 1324-1335.	3.1	7
135	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
136	Calcium-sensing receptor signaling â€" How human disease informs biology. Current Opinion in Endocrine and Metabolic Research, 2021, 16, 10-18.	0.6	7
137	Genetics of monogenic disorders of calcium and bone metabolism. Clinical Endocrinology, 2022, 97, 483-501.	1.2	7
138	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
139	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
140	Control of PTH secretion by the TRPC1 ion channel. JCI Insight, 2020, 5, .	2.3	6
141	Molecular genetic advances in pituitary tumor development. Expert Review of Endocrinology and Metabolism, 2015, 10, 35-53.	1.2	5
142	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
143	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
144	Preclinical drug studies in MEN1-related neuroendocrine neoplasms (MEN1-NENs). Endocrine-Related Cancer, 2020, 27, R345-R355.	1.6	5

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145	Genetic background influences tumour development in heterozygous Men1 knockout mice. Endocrine Connections, 2020, 9, 426-437.	0.8	5
146	Autosomal Dominant Hypocalcemia Type 1 (ADH1) Associated With Myoclonus and Intracerebral Calcifications. Journal of the Endocrine Society, 2022, 6, bvac042.	0.1	5
147	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
148	Pseudohypoparathyroidism type 1a due to a novel mutation in the <scp>GNAS</scp> gene. Clinical Endocrinology, 2016, 84, 463-465.	1.2	4
149	Heterogeneity of G protein activation by the calcium-sensing receptor. Journal of Molecular Endocrinology, 2021, 67, 41-53.	1.1	4
150	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
151	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
152	Calcilytic NPSP795 Increases Plasma Calcium and PTH in an Autosomal Dominant Hypocalcemia Type 1 Mouse Model. JBMR Plus, 2020, 4, e10402.	1.3	3
153	Genetics of Skeletal Disorders. Handbook of Experimental Pharmacology, 2020, 262, 325-351.	0.9	3
154	Studies of mice deleted for Sox3 and uc482: relevance to X-linked hypoparathyroidism. Endocrine Connections, 2020, 9, 173-186.	0.8	3
155	Introduction to Genetics of Skeletal and Mineral Metabolic Diseases. , 2018, , 1-21.		2
156	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
157	Recent advances in calcium-sensing receptor structures and signaling pathways. Progress in Molecular Biology and Translational Science, 2023, , 121-135.	0.9	2
158	SaO021A NOVEL ROLE FOR GATA3 IN MESANGIAL CELLS IN GLOMERULAR DEVELOPMENT AND INJURY. Nephrology Dialysis Transplantation, 2015, 30, iii32-iii32.	0.4	1
159	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
160	The bromodomain inhibitor JQ1+ reduces calcium-sensing receptor activity in pituitary cell lines. Journal of Molecular Endocrinology, 2021, 67, 83-94.	1.1	1
161	GPCR's and Endocrinology. , 2021, , .		1
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