## Sian E Piret

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Factors influencing success of clinical genome sequencing across a broad spectrum of disorders. Nature Genetics, 2015, 47, 717-726.	9.4	310
2	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
3	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
4	ldentification 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
5	ldentification of a G-Protein Subunit-α11 Gain-of-Function Mutation, Val340Met, in a Family With Autosomal Dominant Hypocalcemia Type 2 (ADH2). Journal of Bone and Mineral Research, 2016, 31, 1207-1214.	3.1	36
6	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
7	Mouse model for inherited renal fibrosis associated with endoplasmic reticulum stress. DMM Disease Models and Mechanisms, 2017, 10, 773-786.	1.2	34
8	Krüppel-like factor 6–mediated loss of BCAA catabolism contributes to kidney injury in mice and humans. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	34
9	Mouse models for inherited endocrine and metabolic disorders. Journal of Endocrinology, 2011, 211, 211-230.	1.2	30
10	Gα11 mutation in mice causes hypocalcemia rectifiable by calcilytic therapy. JCI Insight, 2017, 2, e91103.	2.3	28
11	Loss of proximal tubular transcription factor Krüppel-like factor 15 exacerbates kidney injury through loss of fatty acid oxidation. Kidney International, 2021, 100, 1250-1267.	2.6	28
12	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
13	Epidemiology of Uromodulin-Associated Kidney Disease – Results from a Nation-Wide Survey. Nephron Extra, 2012, 2, 147-158.	1.1	25
14	Podocyte-Specific Loss of Krüppel-Like Factor 6 Increases Mitochondrial Injury in Diabetic Kidney Disease. Diabetes, 2018, 67, 2420-2433.	0.3	25
15	Cinacalcet corrects hypercalcemia in mice with an inactivating GÎ $\pm 11$ mutation. JCl Insight, 2017, 2, .	2.3	17
16	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
17	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
18	A Mouse Model of Early-Onset Renal Failure Due to a Xanthine Dehydrogenase Nonsense Mutation. PLoS ONE, 2012, 7, e45217.	1.1	8

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19	Mice with a Brd4 Mutation Represent a New Model of Nephrocalcinosis. Journal of Bone and Mineral Research, 2019, 34, 1324-1335.	3.1	7
20	Genetic background influences tumour development in heterozygous Men1 knockout mice. Endocrine Connections, 2020, 9, 426-437.	0.8	5
21	An <i>N</i> -Ethyl- <i>N</i> -Nitrosourea (ENU)-Induced Tyr265Stop Mutation of the DNA Polymerase Accessory Subunit Gamma 2 ( <i>Polg2</i> ) Is Associated With Renal Calcification in Mice. Journal of Bone and Mineral Research, 2019, 34, 497-507.	3.1	3
22	Studies of mice deleted for Sox3 and uc482: relevance to X-linked hypoparathyroidism. Endocrine Connections, 2020, 9, 173-186.	0.8	3
23	Proximal Tubular Transcription Factors in Acute Kidney Injury: Recent Advances. Nephron, 2020, 144, 613-615.	0.9	2
24	Mouse Models. , 2013, , 181-204.		1
25	An <i>N</i> â€Ethylâ€ <i>N</i> â€Nitrosourea (ENU) Mutagenized Mouse Model for Autosomal Dominant Nonsyndromic Kyphoscoliosis Due to Vertebral Fusion. JBMR Plus, 2018, 2, 154-163.	1.3	1
26	Mouse Models: Approaches to Generate In Vivo Models for Hereditary Disorders of Mineral and Skeletal Homeostasis. , 2018, , 89-118.		0