Judy Savige

List of Publications by Year
in descending order

117 papers	5,007 citations	117571 34 h-index	98753 67 g-index
118 all docs	118 docs citations	118 times ranked	4058 citing authors

#	Article	IF	Citations
1	International Consensus Statement on Testing and Reporting of Antineutrophil Cytoplasmic Antibodies (ANCA). American Journal of Clinical Pathology, 1999, 111, 507-513.	0.4	539
2	Expert Guidelines for the Management of Alport Syndrome and Thin Basement Membrane Nephropathy. Journal of the American Society of Nephrology: JASN, 2013, 24, 364-375.	3.0	285
3	Nonmuscle Myosin Heavy Chain IIA Mutations Define a Spectrum of Autosomal Dominant Macrothrombocytopenias: May-Hegglin Anomaly and Fechtner, Sebastian, Epstein, and Alport-Like Syndromes. American Journal of Human Genetics, 2001, 69, 1033-1045.	2.6	283
4	Array-Based Gene Discovery with Three Unrelated Subjects Shows SCARB2/LIMP-2 Deficiency Causes Myoclonus Epilepsy and Glomerulosclerosis. American Journal of Human Genetics, 2008, 82, 673-684.	2.6	230
5	A review of the ocular manifestations. Ophthalmic Genetics, 1997, 18, 161-173.	0.5	209
6	Thin basement membrane nephropathy. Kidney International, 2003, 64, 1169-1178.	2.6	204
7	Antineutrophil cytoplasmic antibodies and associated diseases: A review of the clinical and laboratory features. Kidney International, 2000, 57, 846-862.	2.6	196
8	Addendum to the International Consensus Statement on Testing and Reporting of Antineutrophil Cytoplasmic Antibodies. American Journal of Clinical Pathology, 2003, 120, 312-318.	0.4	167
9	Ocular Features in Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2015, 10, 703-709.	2.2	147
10	COL4A3/COL4A4 Mutations and Features in Individuals with Autosomal Recessive Alport Syndrome. Journal of the American Society of Nephrology: JASN, 2013, 24, 1945-1954.	3.0	121
11	Mapping structural landmarks, ligand binding sites, and missense mutations to the collagen IV heterotrimers predicts major functional domains, novel interactions, and variation in phenotypes in inherited diseases affecting basement membranes. Human Mutation, 2011, 32, 127-143.	1.1	99
12	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. Pediatric Nephrology, 2019, 34, 1175-1189.	0.9	97
13	Alport Syndrome in Women and Girls. Clinical Journal of the American Society of Nephrology: CJASN, 2016, 11, 1713-1720.	2.2	93
14	Addendum to the International Consensus Statement on Testing and Reporting of Antineutrophil Cytoplasmic Antibodies: Quality Control Guidelines, Comments, and Recommendations for Testing in Other Autoimmune Diseases. American Journal of Clinical Pathology, 2003, 120, 312-318.	0.4	86
15	Prevalence Estimates of Predicted Pathogenic COL4A3â€“COL4A5 Variants in a Population Sequencing Database and Their Implications for Alport Syndrome. Journal of the American Society of Nephrology: JASN, 2021, 32, 2273-2290.	3.0	81
16	Retinal Basement Membrane Abnormalities and the Retinopathy of Alport Syndrome. , 2010, 51, 1621.		80
17	Segregation of hematuria in thin basement membrane disease with haplotypes at the loci for Alport syndrome. Kidney International, 2001, 59, 1670-1676.	2.6	70
18	COL4A4 mutation in thin basement membrane disease previously described in Alport syndrome11See Editorial by Monnens, p. 799. Kidney International, 2001, 60, 480-483.	2.6	70

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19	Mutations in the COL4A4 gene in thin basement membrane disease. Kidney International, 2003, 63, 447-453.	2.6	65
20	The Microvasculature in Chronic Kidney Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2011, 6, 1872-1878.	2.2	65
21	Consensus statement on standards and guidelines for the molecular diagnostics of Alport syndrome: refining the ACMG criteria. European Journal of Human Genetics, 2021, 29, 1186-1197.	1.4	61
22	What do antineutrophil cytoplasmic antibodies (ANCA) tell us?. Best Practice and Research in Clinical Rheumatology, 2005, 19, 263-276.	1.4	58
23	Visual impairment caused by retinal abnormalities in mesangiocapillary (membranoproliferative) glomerulonephritis type II (â€œdense deposit diseaseâ€). American Journal of Kidney Diseases, 2003, 42, e3.1-e3.4.	2.1	56
24	COL4A3 mutations and their clinical consequences in thin basement membrane nephropathy (TBMN)11See Editorial by Gregory, p. 1109.. Kidney International, 2004, 65, 786-790.	2.6	56
25	A comparison of the clinical, histopathologic, and ultrastructural phenotypes in carriers of X-linked and autosomal recessive Alport's syndrome. American Journal of Kidney Diseases, 2001, 38, 1217-1228.	2.1	54
26	Alport syndrome: its effects on the glomerular filtration barrier and implications for future treatment. Journal of Physiology, 2014, 592, 4013-4023.	1.3	51
27	The R229Q mutation in NPHS2 may predispose to proteinuria in thin-basement-membrane nephropathy. Pediatric Nephrology, 2008, 23, 2201-2207.	0.9	49
28	Guidelines for Genetic Testing and Management of Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2022, 17, 143-154.	2.2	49
29	Ocular features aid the diagnosis of Alport syndrome. Nature Reviews Nephrology, 2009, 5, 356-360.	4.1	47
30	NPHS2variation in focal and segmental glomerulosclerosis. BMC Nephrology, 2008, 9, 13.	0.8	46
31	Bull terrier hereditary nephritis: A model for autosomal dominant Alport syndrome. Kidney International, 1995, 47, 758-765.	2.6	45
32	Alport Retinopathy Results from â€œSevereâ€•COL4A5 Mutations and Predicts Early Renal Failure. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 34-38.	2.2	42
33	Stem cell therapy for Alport syndrome: the hope beyond the hype. Nephrology Dialysis Transplantation, 2008, 24, 731-734.	0.4	40
34	Retinal Abnormalities Characteristic of Inherited Renal Disease. Journal of the American Society of Nephrology: JASN, 2011, 22, 1403-1415.	3.0	37
35	Pyoderma gangrenosum with secondary pyarthrosis following propylthiouracil. Australasian Journal of Dermatology, 1999, 40, 144-146.	0.4	35
36	The Genetics of Thin Basement Membrane Nephropathy. Seminars in Nephrology, 2005, 25, 163-170.	0.6	34

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37	Usefulness of N-Terminal Proâ€“Brain Natriuretic Peptide to Predict Postoperative Cardiac Complications and Long-Term Mortality After Emergency Lower Limb Orthopedic Surgery. American Journal of Cardiology, 2010, 106, 865-872.	0.7	34
38	Identifying and integrating consumer perspectives in clinical practice guidelines on autosomalâ€dominant polycystic kidney disease. Nephrology, 2016, 21, 122-132.	0.7	33
39	Antineutrophil cytoplasmic antibody (ANCA)â€positive cutaneous leucocytoclastic vasculitis associated with antithyroid therapy in Graves' disease. Australasian Journal of Dermatology, 1998, 39, 96-99.	0.4	32
40	Clinical and genetic features in autosomal recessive and X-linked Alport syndrome. Pediatric Nephrology, 2014, 29, 391-396.	0.9	32
41	Characterization of the peripheral retinopathy in X-linked and autosomal recessive Alport syndrome. Nephrology Dialysis Transplantation, 2006, 22, 104-108.	0.4	30
42	Nine novel COL4A3 and COL4A4 mutations and polymorphisms identified in inherited membrane diseases. Pediatric Nephrology, 2007, 22, 652-657.	0.9	30
43	The Chemical Chaperone, PBA, Reduces ER Stress and Autophagy and Increases Collagen IV Î±5 Expression in Cultured Fibroblasts From Men With X-Linked Alport Syndrome and Missense Mutations. Kidney International Reports, 2017, 2, 739-748.	0.4	30
44	N-Terminal Pro-Brain Natriuretic Peptide and Angiotensin-Converting Enzyme-2 Levels and Their Association With Postoperative Cardiac Complications After Emergency Orthopedic Surgery. American Journal of Cardiology, 2012, 109, 1365-1373.	0.7	29
45	Does cardiology intervention improve mortality for post-operative troponin elevations after emergency orthopaedicâ€“geriatric surgery? A randomised controlled study. Injury, 2012, 43, 1193-1198.	0.7	29
46	Should We Diagnose Autosomal Dominant Alport Syndrome When There Is a Pathogenic Heterozygous COL4A3 or COL4A4 Variant?. Kidney International Reports, 2018, 3, 1239-1241.	0.4	29
47	Correlation of histopathological features and renal impairment in autosomal dominant Alport syndrome in Bull terriers. Nephrology Dialysis Transplantation, 2002, 17, 1897-1908.	0.4	27
48	Pathogenic Variants in the Genes Affected in Alport Syndrome (COL4A3â€“COL4A5) and Their Association With Other Kidney Conditions: A Review. American Journal of Kidney Diseases, 2021, 78, 857-864.	2.1	27
49	Three novel COL4A4 mutations resulting in stop codons and their clinical effects in autosomal recessive Alport syndrome. Human Mutation, 2002, 20, 321-322.	1.1	26
50	Evaluation of a multiplex flow cytometric immunoassay to detect PR3- and MPO-ANCA in active and treated vasculitis, and in inflammatory bowel disease (IBD). Journal of Immunological Methods, 2008, 336, 104-112.	0.6	26
51	KHAâ€CARI guideline recommendations for the diagnosis and management of autosomal dominant polycystic kidney disease. Nephrology, 2016, 21, 705-716.	0.7	26
52	Antigen-Specific ANCA ELISAs Have Different Sensitivities for Active and Treated Vasculitis and for Nonvasculitic Disease. American Journal of Clinical Pathology, 2008, 129, 42-53.	0.4	25
53	Cardiac injury and troponin testing after orthopaedic surgery. Injury, 2011, 42, 855-863.	0.7	24
54	The collÎ±gen III fibril has a â€œflexi-rodâ€•structure of flexible sequences interspersed with rigid bioactive domains including two with hemostatic roles. PLoS ONE, 2017, 12, e0175582.	1.1	24

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55	Orthopaedicâ€geriatric models of care and their effectiveness. Australasian Journal on Ageing, 2009, 28, 171-176.	0.4	22
56	DNA variant databases improve test accuracy and phenotype prediction in Alport syndrome. Pediatric Nephrology, 2014, 29, 971-977.	0.9	22
57	Clinical, histopathologic, and genetic studies in nine families with focal segmental glomerulosclerosis. American Journal of Kidney Diseases, 2003, 41, 1170-1178.	2.1	21
58	Genotypeâ€“phenotype correlations for COL4A3â€“COL4A5 variants resulting in Gly substitutions in Alport syndrome. Scientific Reports, 2022, 12, 2722.	1.6	21
59	Lupus Anticoagulant in Anti-Neutrophil Cytoplasmic Antibody-Associated Polyarteritis. American Journal of Nephrology, 1995, 15, 157-160.	1.4	20
60	Troponin I and NT-proBNP (N-terminal pro-Brain Natriuretic Peptide) Do Not Predict 6-Month Mortality in Frail Older Patients Undergoing Orthopedic Surgery. Journal of the American Medical Directors Association, 2010, 11, 415-420.	1.2	20
61	Persistent familial hematuria in children and the locus for thin basement membrane nephropathy. Pediatric Nephrology, 2005, 20, 1729-1737.	0.9	19
62	The Risks of Thin Basement Membrane Nephropathy. Seminars in Nephrology, 2005, 25, 171-175.	0.6	19
63	Digenic Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2022, 17, 1697-1706.	2.2	19
64	The use of ocular abnormalities to diagnose X-linked Alport syndrome in children. Pediatric Nephrology, 2008, 23, 1245-1250.	0.9	18
65	Autosomal Dominant Polycystic Kidney Disease: A Path Forward. Seminars in Nephrology, 2015, 35, 524-537.	0.6	18
66	The Epidemiology of Thin Basement Membrane Nephropathy. Seminars in Nephrology, 2005, 25, 136-139.	0.6	15
67	Genotype-Phenotype Correlations for Pathogenic COL4A3â€“COL4A5 Variants in X-Linked, Autosomal Recessive, and Autosomal Dominant Alport Syndrome. Frontiers in Medicine, 2022, 9, .	1.2	15
68	KHA-CARI Autosomal Dominant Polycystic Kidney Disease Guideline: Management of Polycystic Liver Disease. Seminars in Nephrology, 2015, 35, 618-622.e5.	0.6	14
69	Temporal retinal thinning and the diagnosis of Alport syndrome and Thin basement membrane nephropathy. Ophthalmic Genetics, 2018, 39, 208-214.	0.5	14
70	Anti-neutrophil cytoplasmic antibody (ANCA)-associated microscopic polyangiitis following a suppurative wound infection. Nephrology Dialysis Transplantation, 2006, 21, 2993-2994.	0.4	13
71	Testing on formalin-fixed neutrophils is less sensitive and specific for small vessel vasculitis, and less sensitive for MPO-ANCA, than most ELISAs. Journal of Immunological Methods, 2008, 339, 141-145.	0.6	13
72	Most proteinase3â€•and myeloperoxidaseâ€•antineutrophil cytoplasmic antibodies enzymeâ€•linked immunosorbent assays perform less well in treated smallâ€•vessel vasculitis than in active disease. Apmis, 2009, 117, 60-62.	0.9	13

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73	A female with X-linked Alport syndrome and compound heterozygous COL4A5 mutations. Pediatric Nephrology, 2014, 29, 481-485.	0.9	13
74	Retinal Venular Calibre is Increased in Patients with Autoimmune Rheumatic Disease: A Case-Control Study. Current Eye Research, 2013, 38, 685-690.	0.7	12
75	Bullâ€™s eye and pigment maculopathy are further retinal manifestations of an abnormal Bruchâ€™s membrane in Alport syndrome. Ophthalmic Genetics, 2017, 38, 238-244.	0.5	12
76	The 2019 and 2021 International Workshops on Alport Syndrome. European Journal of Human Genetics, 2022, 30, 507-516.	1.4	12
77	Heterozygous Pathogenic COL4A3 and COL4A4 Variants (Autosomal Dominant Alport Syndrome) Are Common, and Not Typically Associated With End-Stage Kidney Failure, Hearing Loss, or Ocular Abnormalities. Kidney International Reports, 2022, 7, 1933-1938.	0.4	12
78	Testing and reporting antineutrophil cytoplasmic antibodies (ANCA) in treated vasculitis and non-vasculitic disease. Journal of Immunological Methods, 2018, 458, 1-7.	0.6	11
79	Microvascular retinopathy and angiographically-demonstrated coronary artery disease: A cross-sectional, observational study. PLoS ONE, 2018, 13, e0192350.	1.1	11
80	KHA-CARI Autosomal Dominant Polycystic Kidney Disease Guideline: Genetic Testing for Diagnosis. Seminars in Nephrology, 2015, 35, 545-549.e2.	0.6	10
81	Hypertensive/Microvascular Disease and COPD: a Case Control Study. Kidney and Blood Pressure Research, 2016, 41, 29-39.	0.9	10
82	Hematuria in Thin Basement Membrane Nephropathy. Seminars in Nephrology, 2005, 25, 146-148.	0.6	9
83	Alport syndrome: deducing the mode of inheritance from the presence of haematuria in family members. Pediatric Nephrology, 2020, 35, 59-66.	0.9	9
84	Microvascular Dilatation after Haemodialysis Is Determined by the Volume of Fluid Removed and Fall in Mean Arterial Pressure. Kidney and Blood Pressure Research, 2012, 35, 644-648.	0.9	8
85	Corneal endothelial cell abnormalities in X-linked Alport syndrome. Ophthalmic Genetics, 2020, 41, 13-19.	0.5	8
86	Do mutations in COL4A1 or COL4A2 cause thin basement membrane nephropathy (TBMN)?. Pediatric Nephrology, 2007, 22, 645-651.	0.9	7
87	Microvascular Disease After Renal Transplantation. Kidney and Blood Pressure Research, 2015, 40, 575-583.	0.9	7
88	Alport Syndrome With Kidney Cysts Is Still Alport Syndrome. Kidney International Reports, 2022, 7, 339-342.	0.4	7
89	Testing for antineutrophil cytoplasmic antibodies. Expert Review of Molecular Diagnostics, 2001, 1, 281-289.	1.5	6
90	KHA-CARI Autosomal Dominant Kidney Disease Guideline: Management of Chronic Pain. Seminars in Nephrology, 2015, 35, 607-611.e3.	0.6	6

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91	Gitelman syndrome and ectopic calcification in the retina and joints. CKJ: Clinical Kidney Journal, 2021, 14, 2023-2028.	1.4	6
92	Thin basement membrane nephropathy and coincidental renal biopsy lesions. Nephrology, 2004, 9, 52-52.	0.7	5
93	Retinal atrophy associated with FSGS in a patient with MELAS syndrome. Kidney International, 2008, 74, 252.	2.6	5
94	Antineutrophil cytoplasmic antibody (ANCA) testing of routine sera varies in different laboratories but concordance is greater for cytoplasmic fluorescence (C-ANCA) and myeloperoxidase specificity (MPO-ANCA). Journal of Immunological Methods, 2009, 347, 19-23.	0.6	5
95	KHA-CARI Autosomal Dominant Polycystic Kidney Disease Guideline: Genetics and Genetic Counseling. Seminars in Nephrology, 2015, 35, 550-556.e1.	0.6	5
96	Gene symbol: COl4A5. Disease: Alport Syndrome. Human Genetics, 2008, 124, 301-2.	1.8	5
97	Ocular clues to the nature of disease causing end-stage renal failure. Nephrology Dialysis Transplantation, 2000, 15, 429-432.	0.4	3
98	A further genetic cause of thin basement membrane nephropathy. Nephrology Dialysis Transplantation, 2016, 31, 1758-1760.	0.4	3
99	Chronic Kidney Disease and Cataract: Seeing the Light. American Journal of Nephrology, 2017, 45, 522-523.	1.4	3
100	Retinal Drusen Are More Common and Larger in Systemic Lupus Erythematosus With Renal Impairment. Kidney International Reports, 2022, 7, 848-856.	0.4	3
101	Retinal drusen in glomerulonephritis with or without immune deposits suggest systemic complement activation in disease pathogenesis. Scientific Reports, 2022, 12, 8234.	1.6	3
102	About timeâ€”treating children with Alport syndrome. Nature Reviews Nephrology, 2012, 8, 375-376.	4.1	2
103	Mutation Databases for Inherited Renal Disease: Are They Complete, Accurate, Clinically Relevant, and Freely Available?. Human Mutation, 2014, 35, 791-793.	1.1	2
104	The Human Variome Project: ensuring the quality of DNA variant databases in inherited renal disease. Pediatric Nephrology, 2015, 30, 1893-1901.	0.9	2
105	Microvascular narrowing and BP monitoring: A single centre observational study. PLoS ONE, 2019, 14, e0210625.	1.1	2
106	Increased retinal venular calibre in acute infections. Scientific Reports, 2021, 11, 17280.	1.6	2
107	Pathogenic LAMA5 Variants and Kidney Disease. Kidney360, 2021, 2, 1876-1879.	0.9	2
108	Asymptomatic microscopic hematuria. Urology, 2002, 59, 631-632.	0.5	1

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109	Increased microvascular disease in X-linked and autosomal recessive Alport syndrome: a case control cross sectional observational study. Ophthalmic Genetics, 2019, 40, 129-134.	0.5	1
110	Identification of 27 Novel Variants in Genes COL4A3, COL4A4, and COL4A5 in Lithuanian Families With Alport Syndrome. Frontiers in Medicine, 2022, 9, 859521.	1.2	1
111	RE: THE ASSOCIATION OF AN INCREASED URINARY CALCIUM-TO-CREATININE RATIO, AND ASYMPTOMATIC GROSS AND MICROSCOPIC HEMATURIA IN CHILDREN. Journal of Urology, 2002, 168, 2126-2127.	0.2	0
112	Acute Glomerulonephritis. , 2019, , 275-282.e2.		0
113	LACK OF SEGREGATION OF HAEMATURIA IN THIN BASEMENT MEMBRANE DISEASE (TBMD) WITH HAPLOTYPES AT THE LOCI FOR NIDOGEN, PERLECAN, FIBRONECTIN AND LAMININ. Nephrology, 2000, 5, A94-A94.	0.7	0
114	RE: THE ASSOCIATION OF AN INCREASED URINARY CALCIUM-TO-CREATININE RATIO, AND ASYMPTOMATIC GROSS AND MICROSCOPIC HEMATURIA IN CHILDREN. Journal of Urology, 2002, , 2126-2127.	0.2	0
115	Poststreptococcal Glomerulonephritis. , 2009, , 855-859.		0
116	Gene symbol: COL4A5. Disease: Alport Syndrome. Human Genetics, 2008, 124, 301.	1.8	0
117	Gene symbol: COl4A5. Disease: Alport Syndrome. Human Genetics, 2008, 124, 302.	1.8	0