Tomohiko Yamamura

List of Publications by Year
in descending order

70	1,255	471509 17	434195 31
papers	citations	h-index	g-index

71	71	71	1274
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical features of autosomal recessive polycystic kidney disease in the Japanese population and analysis of splicing in PKHD1 gene for determination of phenotypes. Clinical and Experimental Nephrology, 2022, 26, 140-153.	1.6	2
2	Last Nucleotide Substitutions of COL4A5 Exons Cause Aberrant Splicing. Kidney International Reports, 2022, 7, 108-116.	0.8	14
3	BCS1L mutations produce Fanconi syndrome with developmental disability. Journal of Human Genetics, 2022, 67, 143-148.	2.3	5
4	Evaluation of suspected autosomal Alport Syndrome synonymous variants. Kidney360, 2022, 3, 10.34067/KID.0005252021.	2.1	3
5	Use of renin-angiotensin system inhibitors as initial therapy in children with Henochâ€“SchÃ¶nlein purpura nephritis of moderate severity. Pediatric Nephrology, 2022, 37, 1845-1853.	1.7	5
6	Detecting MUC1 Variants in Patients Clinicopathologically Diagnosed With Having Autosomal Dominant Tubulointerstitial Kidney Disease. Kidney International Reports, 2022, 7, 857-866.	0.8	7
7	Efficacy of combination therapy for childhood complicated focal IgA nephropathy. Clinical and Experimental Nephrology, 2022, , 1.	1.6	1
8	The Contribution of COL4A5 Splicing Variants to the Pathogenesis of X-Linked Alport Syndrome. Frontiers in Medicine, 2022, 9, 841391.	2.6	10
9	Comprehensive genetic analysis using next-generation sequencing for the diagnosis of nephronophthisis-related ciliopathies in the Japanese population. Journal of Human Genetics, 2022, 67, 427-440.	2.3	5
10	Mycophenolate Mofetil after Rituximab for Childhood-Onset Complicated Frequently-Relapsing or Steroid-Dependent Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2022, 33, 401-419.	6.1	24
11	FAT1 biallelic truncating mutation causes a non-syndromic proteinuria in a child. CEN Case Reports, 2021, 10, 100-105.	0.9	3
12	Usefulness of functional splicing analysis to confirm precise disease pathogenesis in Diamond-Blackfan anemia caused by intronic variants in RPS19. Pediatric Hematology and Oncology, 2021, 38, 515-527.	0.8	3
13	Utility of glomerular Gd-IgA1 staining for indistinguishable cases of IgA nephropathy or Alport syndrome. Clinical and Experimental Nephrology, 2021, 25, 779-787.	1.6	1
14	Clinical and histological features in pediatric and adolescent/young adult patients with renal disease: a cross-sectional analysis of the Japan Renal Biopsy Registry (J-RBR). Clinical and Experimental Nephrology, 2021, 25, 1018-1026.	1.6	3
15	X-chromosome inactivation patterns in females with Fabry disease examined by both ultra-deep RNA sequencing and methylation-dependent assay. Clinical and Experimental Nephrology, 2021, 25, 1224-1230.	1.6	9
16	Systematic Review of Genotype-Phenotype Correlations in Frasier Syndrome. Kidney International Reports, 2021, 6, 2585-2593.	0.8	12
17	Genotype-Phenotype Correlation in WT1 Exon 8 to 9 Missense Variants. Kidney International Reports, 2021, 6, 2114-2121.	0.8	7
18	Examination of the predicted prevalence of Gitelman syndrome by ethnicity based on genome databases. Scientific Reports, 2021, 11, 16099.	3.3	13

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19	Identification of novel OCRL isoforms associated with phenotypic differences between Dent disease-2 and Lowe syndrome. Nephrology Dialysis Transplantation, 2021, , .	0.7	0
20	A disease-causing variant of COL4A5 in a Chinese family with Alport syndrome: a case series. BMC Nephrology, 2021, 22, 380.	1.8	2
21	Both the rituximab dose and maintenance immunosuppression in steroid-dependent/frequently-relapsing nephrotic syndrome have important effects on outcomes. Kidney International, 2020, 97, 393-401.	5.2	69
22	Inherited saltâ€losing tubulopathy: An old condition but a new category of tubulopathy. Pediatrics International, 2020, 62, 428-437.	0.5	16
23	Comparison of clinical and genetic characteristics between Dent disease 1 and Dent disease 2. Pediatric Nephrology, 2020, 35, 2319-2326.	1.7	5
24	Genotype-phenotype correlations influence the response to angiotensin-targeting drugs in Japanese patients with male X-linked Alport syndrome. Kidney International, 2020, 98, 1605-1614.	5.2	55
25	A case with somatic and germline mosaicism in COL4A5 detected by multiplex ligation-dependent probe amplification in X-linked Alport syndrome. CEN Case Reports, 2020, 9, 431-436.	0.9	1
26	Glomerular galactose-deficient IgA1 expression analysis in pediatric patients with glomerular diseases. Scientific Reports, 2020, 10, 14026.	3.3	8
27	Development of an exon skipping therapy for X-linked Alport syndrome with truncating variants in COL4A5. Nature Communications, 2020, 11, 2777.	12.8	46
28	Trimerization and Genotypeâ€“Phenotype Correlation of COL4A5 Mutants in Alport Syndrome. Kidney International Reports, 2020, 5, 718-726.	0.8	18
29	Pathogenic evaluation of synonymous COL4A5 variants in Xâ€linked Alport syndrome using a minigene assay. Molecular Genetics & Genomic Medicine, 2020, 8, e1342.	1.2	16
30	Common risk variants in NPHS1 and TNFSF15 are associated with childhood steroid-sensitive nephrotic syndrome. Kidney International, 2020, 98, 1308-1322.	5.2	39
31	Clinical and genetic variability of PAX2-related disorder in the Japanese population. Journal of Human Genetics, 2020, 65, 541-549.	2.3	15
32	Functional analysis of suspected splicing variants in CLCN5 gene in Dent disease 1. Clinical and Experimental Nephrology, 2020, 24, 606-612.	1.6	9
33	Poststreptococcal acute glomerulonephritis can be a risk factor for accelerating kidney dysfunction in Alport syndrome: a case experience. CEN Case Reports, 2020, 9, 418-422.	0.9	2
34	Molecular mechanisms determining severity in patients with Pierson syndrome. Journal of Human Genetics, 2020, 65, 355-362.	2.3	8
35	Comprehensive genetic diagnosis of Japanese patients with severe proteinuria. Scientific Reports, 2020, 10, 270.	3.3	50
36	Genetic background, recent advances in molecular biology, and development of novel therapy in Alport syndrome. Kidney Research and Clinical Practice, 2020, 39, 402-413.	2.2	13

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37	Heterozygous Urinary Abnormalityâ€“Causing Variants of COL4A3 and COL4A4 Affect Severity of Autosomal Recessive Alport Syndrome. Kidney360, 2020, 1, 936-942.	2.1	2
38	A review of clinical characteristics and genetic backgrounds in Alport syndrome. Clinical and Experimental Nephrology, 2019, 23, 158-168.	1.6	135
39	Comparison between conventional and comprehensive sequencing approaches for genetic diagnosis of Alport syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e883.	1.2	25
40	Determination of the pathogenicity of known COL4A5 intronic variants by in vitro splicing assay. Scientific Reports, 2019, 9, 12696.	3.3	14
41	Clinical characteristics of HNF1B-related disorders in a Japanese population. Clinical and Experimental Nephrology, 2019, 23, 1119-1129.	1.6	31
42	Molecular assay for an intronic variant in NUP93 that causes steroid resistant nephrotic syndrome. Journal of Human Genetics, 2019, 64, 673-679.	2.3	12
43	TGFBI-associated corneal dystrophy and nephropathy: a novel syndrome?. CEN Case Reports, 2019, 8, 14-17.	0.9	3
44	Clinical spectrum of male patients with OFD1 mutations. Journal of Human Genetics, 2019, 64, 3-9.	2.3	12
45	Clinical and Genetic Characteristics in Patients With Gitelman Syndrome. Kidney International Reports, 2019, 4, 119-125.	0.8	47
46	Pair analysis and custom array CGH can detect a small copy number variation in COQ6 gene. Clinical and Experimental Nephrology, 2019, 23, 669-675.	1.6	8
47	Development of ultra-deep targeted RNA sequencing for analyzing X-chromosome inactivation in female Dent disease. Journal of Human Genetics, 2018, 63, 589-595.	2.3	10
48	Detection of copy number variations by pair analysis using next-generation sequencing data in inherited kidney diseases. Clinical and Experimental Nephrology, 2018, 22, 881-888.	1.6	25
49	Functional splicing analysis in an infantile case of atypical hemolytic uremic syndrome caused by digenic mutations in C3 and MCP genes. Journal of Human Genetics, 2018, 63, 755-759.	2.3	7
50	Detection of a Splice Site Variant in a Patient with Glomerulopathy and Fibronectin Deposits. Nephron, 2018, 138, 166-171.	1.8	5
51	The utility of urinary CD80 as a diagnostic marker in patients with renal diseases. Scientific Reports, 2018, 8, 17322.	3.3	12
52	Three Severe Cases of Viral Infections with Post-Kidney Transplantation Successfully Confirmed by Polymerase Chain Reaction and Flow Cytometry. Case Reports in Nephrology and Dialysis, 2018, 8, 198-206.	0.6	1
53	Congenital chloride diarrhea needs to be distinguished from Bartter and Gitelman syndrome. Journal of Human Genetics, 2018, 63, 887-892.	2.3	20
54	Detection of Splicing Abnormalities and Genotype-Phenotype Correlation in X-linked Alport Syndrome. Journal of the American Society of Nephrology: JASN, 2018, 29, 2244-2254.	6.1	43

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55	Rare renal ciliopathies in non-consanguineous families that were identified by targeted resequencing. Clinical and Experimental Nephrology, 2017, 21, 136-142.	1.6	9
56	Clinical characteristics and long-term outcome of diarrhea-associated hemolytic uremic syndrome: a single center experience. Clinical and Experimental Nephrology, 2017, 21, 889-894.	1.6	10
57	Natural History and Genotypeâ€“Phenotype Correlation in Female X-Linked Alport Syndrome. Kidney International Reports, 2017, 2, 850-855.	0.8	62
58	Diagnostic strategy for inherited hypomagnesemia. Clinical and Experimental Nephrology, 2017, 21, 1003-1010.	1.6	8
59	Characterization of contiguous gene deletions in COL4A6 and COL4A5 in Alport syndrome-diffuse leiomyomatosis. Journal of Human Genetics, 2017, 62, 733-735.	2.3	20
60	A birth of bipartite exon by intragenic deletion. Molecular Genetics & Genomic Medicine, 2017, 5, 287-294.	1.2	4
61	A comparison of splicing assays to detect an intronic variant of the OCRL gene in Lowe syndrome. European Journal of Medical Genetics, 2017, 60, 631-634.	1.3	15
62	Changes in the numbers of patients with acute gastroenteritis after voluntary introduction of the rotavirus vaccine in a Japanese childrenâ€™s primary emergency medical center. Environmental Health and Preventive Medicine, 2017, 22, 15.	3.4	5
63	Female X-linked Alport syndrome with somatic mosaicism. Clinical and Experimental Nephrology, 2017, 21, 877-883.	1.6	13
64	Cryptic exon activation in SLC12A3 in Gitelman syndrome. Journal of Human Genetics, 2017, 62, 335-337.	2.3	12
65	An in vitro splicing assay reveals the pathogenicity of a novel intronic variant in ATP6V0A4 for autosomal recessive distal renal tubular acidosis. BMC Nephrology, 2017, 18, 353.	1.8	10
66	A Novel Mutation in a Japanese Family with X-linked Alport Syndrome. Internal Medicine, 2016, 55, 2843-2847.	0.7	4
67	Genetic, Clinical, and Pathologic Backgrounds of Patients with Autosomal Dominant Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2016, 11, 1441-1449.	4.5	94
68	Pathogenesis of hypokalemia in autosomal dominant hypocalcemia type 1. Clinical and Experimental Nephrology, 2016, 20, 253-257.	1.6	8
69	Differential diagnosis of Bartter syndrome, Gitelman syndrome, and pseudoâ€“Bartter/Gitelman syndrome based on clinical characteristics. Genetics in Medicine, 2016, 18, 180-188.	2.4	67
70	Detecting pathogenic deep intronic variants in <scp>Gitelman</scp> syndrome. American Journal of Medical Genetics, Part A, 0, , .	1.2	3