Gianluca Caridi

List of Publications by Year
in descending order

159	7,244	66343	64796
papers	citations	42 h-index	79 g-index

163	163	163	7963
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Discovery of new risk loci for IgA nephropathy implicates genes involved in immunity against intestinal pathogens. Nature Genetics, 2014, 46, 1187-1196.	21.4	505
2	Mutations in MYH9 result in the May-Hegglin anomaly, and Fechtner and Sebastian syndromes. Nature Genetics, 2000, 26, 103-105.	21.4	397
3	Renal outcome in patients with congenital anomalies of the kidney and urinary tract. Kidney International, 2009, 76, 528-533.	5.2	309
4	COQ2 Nephropathy. Journal of the American Society of Nephrology: JASN, 2007, 18, 2773-2780.	6.1	297
5	IgA nephropathy, the most common cause of glomerulonephritis, is linked to 6q22â€“23. Nature Genetics, 2000, 26, 354-357.	21.4	291
6	Allelism of MCKD, FJHN and GCKD caused by impairment of uromodulin export dynamics. Human Molecular Genetics, 2003, 12, 3369-3384.	2.9	203
7	Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. American Journal of Human Genetics, 2012, 91, 987-997.	6.2	201
8	AHI1 is required for photoreceptor outer segment development and is a modifier for retinal degeneration in nephronophthisis. Nature Genetics, 2010, 42, 175-180.	21.4	171
9	Broadening the Spectrum of Diseases Related to Podocin Mutations. Journal of the American Society of Nephrology: JASN, 2003, 14, 1278-1286.	6.1	159
10	Prevalence, Genetics, and Clinical Features of Patients Carrying Podocin Mutations in Steroid-Resistant Nonfamilial Focal Segmental Glomerulosclerosis. Journal of the American Society of Nephrology: JASN, 2001, 12, 2742-2746.	6.1	155
11	Whole-Exome Sequencing in Adults With Chronic Kidney Disease. Annals of Internal Medicine, 2018, 168, 100.	3.9	154
12	Recurrence of focal segmental glomerulosclerosis after renal transplantation in patients with mutations of podocin. American Journal of Kidney Diseases, 2003, 41, 1314-1321.	1.9	144
13	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	6.2	137
14	Uromodulin storage diseases: Clinical aspects and mechanisms. American Journal of Kidney Diseases, 2004, 44, 987-999.	1.9	123
15	Mutations in DSTYK and Dominant Urinary Tract Malformations. New England Journal of Medicine, 2013, 369, 621-629.	27.0	119
16	Genotypeâ€“phenotype associations in WT1 glomerulopathy. Kidney International, 2014, 85, 1169-1178.	5.2	113
17	Rituximab is a safe and effective long-term treatment for children with steroid and calcineurin inhibitorâ€“dependent idiopathic nephrotic syndrome. Kidney International, 2013, 84, 1025-1033.	5.2	109
18	Identification of a New Locus for Medullary Cystic Disease, on Chromosome 16p12. American Journal of Human Genetics, 1999, 64, 1655-1660.	6.2	104

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19	Genetic approaches to human renal agenesis/hypoplasia and dysplasia. Pediatric Nephrology, 2007, 22, 1675-1684.	1.7	99
20	CD2AP mutations are associated with sporadic nephrotic syndrome and focal segmental glomerulosclerosis (FSGS). Nephrology Dialysis Transplantation, 2009, 24, 1858-1864.	0.7	97
21	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	2.5	96
22	Genetics, clinical and pathological features of glomerulonephrites associated with mutations of nonmuscle myosin IIA (Fechtner syndrome). American Journal of Kidney Diseases, 2003, 41, 95-104.	1.9	94
23	Renal-retinal syndromes: Association of retinal anomalies and recessive nephronophthisis in patients with homozygous deletion of the NPH1 locus. American Journal of Kidney Diseases, 1998, 32, 1059-1062.	1.9	93
24	TRPC6 Mutations in Children with Steroid-Resistant Nephrotic Syndrome and Atypical Phenotype. Clinical Journal of the American Society of Nephrology: CJASN, 2011, 6, 1626-1634.	4.5	89
25	NPHS2 (Podocin) Mutations in Nephrotic Syndrome. Clinical Spectrum and Fine Mechanisms. Pediatric Research, 2005, 57, 54R-61R.	2.3	85
26	Active Focal Segmental Glomerulosclerosis Is Associated with Massive Oxidation of Plasma Albumin. Journal of the American Society of Nephrology: JASN, 2007, 18, 799-810.	6.1	83
27	Serum Glomerular Permeability Activity in Patients with Podocin Mutations (NPHS2) and Steroid-ResistantNephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2002, 13, 1946-1952.	6.1	77
28	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
29	Genetic screening in adolescents with steroid-resistant nephrotic syndrome. Kidney International, 2013, 84, 206-213.	5.2	77
30	Anti-CD20 Antibodies for Idiopathic Nephrotic Syndrome in Children. Clinical Journal of the American Society of Nephrology: CJASN, 2016, 11, 710-720.	4.5	70
31	Exome sequencing identified MYO1E and NEIL1 as candidate genes for human autosomal recessive steroid-resistant nephrotic syndrome. Kidney International, 2011, 80, 389-396.	5.2	69
32	Induction of apoptosis in human neuroblastoma cells by abrogation of integrin-mediated cell adhesion. International Journal of Cancer, 1997, 70, 688-698.	5.1	68
33	RPGRIP1L mutations are mainly associated with the cerebelloâ€renal phenotype of Joubert syndromeâ€related disorders. Clinical Genetics, 2008, 74, 164-170.	2.0	64
34	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
35	Mutation of the Mg2+ Transporter SLC41A1 Results in a Nephronophthisis-Like Phenotype. Journal of the American Society of Nephrology: JASN, 2013, 24, 967-977.	6.1	63
36	Familial Vesicoureteral Reflux: Testing Replication of Linkage in Seven New Multigenerational Kindreds. Journal of the American Society of Nephrology: JASN, 2005, 16, 1781-1787.	6.1	56

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37	Depletion of clusterin in renal diseases causing nephrotic syndrome. Kidney International, 2002, 62, 2184-2194.	5.2	55
38	Mutations in SOX17 are associated with congenital anomalies of the kidney and the urinary tract. Human Mutation, 2010, 31, 1352-1359.	2.5	54
39	Podocin mutations in sporadic focal-segmental glomerulosclerosis occurring in adulthood. Kidney International, 2003, 64, 365.	5.2	53
40	Cyclosporine in patients with steroid-resistant nephrotic syndrome: an open-label, nonrandomized, retrospective study. Clinical Therapeutics, 2004, 26, 1411-1418.	2.5	48
41	WT1 mutations in nephrotic syndrome revisited. High prevalence in young girls, associations and renal phenotypes. Pediatric Nephrology, 2006, 21, 1393-1398.	1.7	46
42	Adverse events linked with the use of chimeric and humanized antiâ€CD20 antibodies in children with idiopathic nephrotic syndrome. British Journal of Clinical Pharmacology, 2018, 84, 1238-1249.	2.4	46
43	A Recessive Gene for Primary Vesicoureteral Reflux Maps to Chromosome 12p11-q13. Journal of the American Society of Nephrology: JASN, 2009, 20, 1633-1640.	6.1	42
44	Familial forms of nephrotic syndrome. Pediatric Nephrology, 2010, 25, 241-252.	1.7	41
45	A novel hepatocyte nuclear factor-1Î² (MODY-5) gene mutation in an Italian family with renal dysfunctions and early-onset diabetes. Diabetologia, 2002, 45, 153-154.	6.3	39
46	Nephronophthisis type 1 deletion syndrome with neurological symptoms: Prevalence and significance of the association. Kidney International, 2006, 70, 1342-1347.	5.2	39
47	Clinical Features and Long-Term Outcome of Nephrotic Syndrome Associated with Heterozygous NPHS1 and NPHS2 Mutations. Clinical Journal of the American Society of Nephrology: CJASN, 2009, 4, 1065-1072.	4.5	38
48	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
49	Molecular and Cellular Mechanisms for Proteinuria in Minimal Change Disease. Frontiers in Medicine, 2018, 5, 170.	2.6	38
50	Congenital analbuminaemia: Molecular defects and biochemical and clinical aspects. Biochimica Et Biophysica Acta - General Subjects, 2013, 1830, 5494-5502.	2.4	37
51	crv4, a mouse model for human ataxia associated with kyphoscoliosis caused by an mRNA splicing mutation of the metabotropic glutamate receptor 1 (Grm1). International Journal of Molecular Medicine, 2006, 18, 593-600.	4.0	36
52	Clinical and molecular heterogeneity of juvenile nephronophthisis in Italy: Insights from molecular screening. American Journal of Kidney Diseases, 2000, 35, 44-51.	1.9	34
53	Effects of peritoneal effluents on mesothelial cells m culture: cell proliferation and extracellular matrix regulation. Nephrology Dialysis Transplantation, 1996, 11, 1803-1809.	0.7	33
54	Localization of a Gene for Nonsyndromic Renal Hypodysplasia to Chromosome 1p32-33. American Journal of Human Genetics, 2007, 80, 539-549.	6.2	33

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55	Collapsing glomerulopathy associated with inherited mitochondrial injury. Kidney International, 2008, 74, 237-243.	5.2	31
56	Albuminuria and Glomerular Damage in Mice Lacking the Metabotropic Glutamate Receptor 1. American Journal of Pathology, 2011, 178, 1257-1269.	3.8	31
57	Locus heterogeneity of Dentâ€™s disease: OCRL1 and TMEM27 genes in patients with no CLCN5 mutations. Pediatric Nephrology, 2009, 24, 1967-1973.	1.7	30
58	A Novel WT1 Gene Mutation in a Three-Generation Family with Progressive Isolated Focal Segmental Glomerulosclerosis. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 698-702.	4.5	30
59	COVID-19 in Children with Nephrotic Syndrome on Anti-CD20 Chronic Immunosuppression. Clinical Journal of the American Society of Nephrology: CJASN, 2020, 15, 1494-1495.	4.5	30
60	Genetic and Clinical Predictors of Age of ESKD in Individuals With Autosomal Dominant Tubulointerstitial Kidney Disease Due to UMOD Mutations. Kidney International Reports, 2020, 5, 1472-1485.	0.8	30
61	Cell-specific regulation of Â1(III) and Â2(V) collagen by TGF-Â1 in tubulointerstitial cell models. Nephrology Dialysis Transplantation, 1998, 13, 573-579.	0.7	29
62	Novel INF2 mutations in an Italian cohort of patients with focal segmental glomerulosclerosis, renal failure and Charcot-Marie-Tooth neuropathy. Nephrology Dialysis Transplantation, 2014, 29, iv80-iv86.	0.7	28
63	Post-transplant recurrence of steroid resistant nephrotic syndrome in children: the Italian experience. Journal of Nephrology, 2020, 33, 849-857.	2.0	28
64	RORB gene and 9q21.13 microdeletion: Report on a patient with epilepsy and mild intellectual disability. European Journal of Medical Genetics, 2014, 57, 44-46.	1.3	26
65	Type IV Collagen Mutations in Familial IgA Nephropathy. Kidney International Reports, 2020, 5, 1075-1078.	0.8	26
66	Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801.	1.1	26
67	Autosomal dominant medullary cystic disease: a disorder with variable clinical pictures and exclusion of linkage with the NPH1 locus. Nephrology Dialysis Transplantation, 1998, 13, 2536-2546.	0.7	25
68	Analbuminemia Produced by a Novel Splicing Mutation. Clinical Chemistry, 2007, 53, 1549-1552.	3.2	25
69	Association of the macrophage migration inhibitory factor â^'173*C allele with childhood nephrotic syndrome. Pediatric Nephrology, 2008, 23, 743-748.	1.7	25
70	Glomerulocystic kidney disease in a family. Nephrology Dialysis Transplantation, 2002, 17, 813-818.	0.7	24
71	Further phenotypic heterogeneity of <scp>CoQ10</scp> deficiency associated with steroid resistant nephrotic syndrome and novel <scp>COQ2</scp> and <scp>COQ6</scp> variants. Clinical Genetics, 2017, 92, 224-226.	2.0	24
72	Apolipoprotein E in idiopathic nephrotic syndrome and focal segmental glomerulosclerosis. Kidney International, 2003, 63, 686-695.	5.2	23

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73	Immature Renal Structures Associated With a Novel UMOD Sequence Variant. American Journal of Kidney Diseases, 2009, 53, 327-331.	1.9	23
74	Mutational Spectrum of CYP24A1 Gene in a Cohort of Italian Patients with Idiopathic Infantile Hypercalcemia. Nephron, 2016, 133, 193-204.	1.8	23
75	Infantile steroid-resistant nephrotic syndrome associated with double homozygous mutations of podocin. American Journal of Kidney Diseases, 2004, 43, 727-732.	1.9	22
76	Genetic risk factors in typical haemolytic uraemic syndrome. Nephrology Dialysis Transplantation, 2009, 24, 1851-1857.	0.7	22
77	Diagnosis, Phenotype, and Molecular Genetics of Congenital Analbuminemia. Frontiers in Genetics, 2019, 10, 336.	2.3	22
78	Expression of Nuclear Transcription Factor PAX2 in Renal Biopsies of Juvenile Nephronophthisis. Nephron, 2002, 91, 588-593.	1.8	21
79	Rituximab vs Low-Dose Mycophenolate Mofetil in Recurrence of Steroid-Dependent Nephrotic Syndrome in Children and Young Adults. JAMA Pediatrics, 2021, 175, 631.	6.2	21
80	Evidence of further genetic heterogeneity in autosomal dominant medullary cystic kidney disease. Nephrology Dialysis Transplantation, 2000, 15, 818-821.	0.7	20
81	Urinary secretion and extracellular aggregation of mutant uromodulin isoforms. Kidney International, 2012, 81, 769-778.	5.2	20
82	A Retrotransposon Insertion in the 5â€² Regulatory Domain of Ptf1a Results in Ectopic Gene Expression and Multiple Congenital Defects in Danforth's Short Tail Mouse. PLoS Genetics, 2013, 9, e1003206.	3.5	20
83	Posttransplant Recurrence of Proteinuria in a Case of Focal Segmental Glomerulosclerosis Associated with WT1 Mutation. American Journal of Transplantation, 2006, 6, 2208-2211.	4.7	19
84	Autosomal Dominant Tubulointerstitial Kidney Disease with Adult Onset due to a Novel Renin Mutation Mapping in the Mature Protein. Scientific Reports, 2019, 9, 11601.	3.3	19
85	Low-dose ofatumumab for multidrug-resistant nephrotic syndrome in children: a randomized placebo-controlled trial. Pediatric Nephrology, 2020, 35, 997-1003.	1.7	19
86	Homocysteine, folate, vitamin B12 levels, and C677T MTHFR mutation in children with renal failure. Pediatric Nephrology, 2003, 18, 225-229.	1.7	18
87	A novel mutation in the albumin gene (c.1A>C) resulting in analbuminemia. European Journal of Clinical Investigation, 2013, 43, 72-78.	3.4	18
88	The First Intron of the Human Osteopontin Gene Contains a C/EBP-Beta-Responsive Enhancer. Gene Expression, 2003, 11, 95-104.	1.2	18
89	Direct effect of plasma permeability factors from patients with idiopatic FSGS on nephrin and podocin expression in human podocytes. International Journal of Molecular Medicine, 2005, 16, 49-58.	4.0	18
90	Improved strategy for molecular genetic diagnostics in juvenile nephronophthisis. American Journal of Kidney Diseases, 2001, 37, 1131-1139.	1.9	17

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91	Cis and trans regulatory elements in NPHS2 promoter: Implications in proteinuria and progression of renal diseases. Kidney International, 2006, 70, 1332-1341.	5.2	16
92	Novel HAX1 gene mutations associated to neurodevelopment abnormalities in two Italian patients with severe congenital neutropenia. Haematologica, 2010, 95, 168-169.	3.5	16
93	Mapping of the human COL5A1 gene to chromosome 9q34.3. Human Genetics, 1992, 90, 174-6.	3.8	15
94	Extracellular matrix formation by epithelial cells from human polycystic kidney cysts in culture. Vigiliae Christianae, 1993, 63, 1-9.	0.1	15
95	A DNA element in the Î±1 type III collagen promoter mediates a stimulatory response by angiotensin II. Kidney International, 2000, 58, 537-548.	5.2	15
96	Glomerular albumin permeability as anin vitromodel for characterizing the mechanism of focal glomerulosclerosis and predicting post-transplant recurrence. Pediatric Transplantation, 2004, 8, 339-343.	1.0	15
97	Use of Anticoagulants and Antiplatelet Agents in Stable Outpatients with Coronary Artery Disease and Atrial Fibrillation. International CLARIFY Registry. PLoS ONE, 2015, 10, e0125164.	2.5	15
98	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. Nephrology Dialysis Transplantation, 2020, 35, 1195-1202.	0.7	15
99	A novel frameshift deletion in the albumin gene causes analbuminemia in a young Turkish woman. Clinica Chimica Acta, 2010, 411, 1711-1715.	1.1	14
100	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. American Journal of Human Genetics, 2021, 108, 357-367.	6.2	14
101	Nephronophthisis-medullary cystic disease: clinical and genetic aspects. Journal of Nephrology, 1998, 11, 224-8.	2.0	14
102	Apoptosis of Human Neuroblastoma Cells Induced by Liposome-Encapsulated Fenretinide. Journal of Liposome Research, 1998, 8, 401-423.	3.3	13
103	A novel splicing mutation causes analbuminemia in a Portuguese boy. Molecular Genetics and Metabolism, 2012, 105, 479-483.	1.1	13
104	Rare Functional Variants of Podocin (NPHS2) Promoter in Patients With Nephrotic Syndrome. Gene Expression, 2006, 13, 59-66.	1.2	12
105	Analbuminemia Zonguldak: Case report and mutational analysis. Clinical Biochemistry, 2008, 41, 288-291.	1.9	12
106	Effects of peritoneal effluents on mesothelial cells in culture: cell proliferation and extracellular matrix regulation. Nephrology Dialysis Transplantation, 1996, 11, 1803-9.	0.7	12
107	Lack of cardiac anomalies in children with NPHS2 mutations. Nephrology Dialysis Transplantation, 2007, 22, 1477-1479.	0.7	11
108	Molecular Diagnosis of Analbuminemia: A New Case Caused by a Nonsense Mutation in the Albumin Gene. International Journal of Molecular Sciences, 2011, 12, 7314-7322.	4.1	11

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109	Localisation of the gene responsible for Fechtner syndrome in a region <600 Kb on 22q11â€“q13. European Journal of Human Genetics, 2000, 8, 895-899.	2.8	10
110	Glomerulocystic kidney disease in hypomelanosis of Ito. Pediatric Nephrology, 2008, 23, 1183-1187.	1.7	10
111	Five cases of severe vesico-ureteric reflux in a family with an X-linked compatible trait. Pediatric Nephrology, 2010, 25, 349-352.	1.7	10
112	A novel frameâ€shift deletion causing analbuminaemia in an Italian paediatric patient. European Journal of Clinical Investigation, 2010, 40, 281-284.	3.4	10
113	Congenital analbuminemia caused by a novel aberrant splicing in the albumin gene. Biochemia Medica, 2014, 24, 151-158.	2.7	10
114	crv4, a mouse model for human ataxia associated with kyphoscoliosis caused by an mRNA splicing mutation of the metabotropic glutamate receptor 1 (Grm1). International Journal of Molecular Medicine, 2006, 18, 593.	4.0	9
115	Analbuminemia in a Swedish male is caused by the Kayseri mutation (c228_229delAT). Clinica Chimica Acta, 2008, 396, 89-92.	1.1	9
116	Urine proteome analysis in Dent's disease shows high selective changes potentially involved in chronic renal damage. Journal of Proteomics, 2016, 130, 26-32.	2.4	9
117	Medullary Cystic Kidney Disease: Past and Present. , 2001, 136, 68-78.		8
118	Heterozygous NPHS1 or NPHS2 mutations in responsive nephrotic syndrome and the multifactorial origin of proteinuria. Kidney International, 2004, 66, 1715-1716.	5.2	8
119	Congenital analbuminaemia diagnosed in adulthood in an Australian family. Pathology, 2015, 47, 492-494.	0.6	8
120	Variations in the Human Serum Albumin Gene: Molecular and Functional Aspects. International Journal of Molecular Sciences, 2022, 23, 1159.	4.1	8
121	Molecular analysis of uromodulin and SAH genes, positional candidates for autosomal dominant medullary cystic kidney disease linked to 16p12. Journal of Nephrology, 2001, 14, 392-6.	2.0	8
122	Direct effect of plasma permeability factors from patients with idiopatic FSGS on nephrin and podocin expression in human podocytes. International Journal of Molecular Medicine, 2005, 16, 49.	4.0	7
123	Podocin-Related Mechanisms in Posttransplantation Recurrence of Focal Segmental Glomerulsclerosis. Transplantation Proceedings, 2006, 38, 3486-3490.	0.6	7
124	A novel nonsense mutation in the albumin gene (c.1275 C>A) causing analbuminemia in a Tunisian boy. Clinical Chemistry and Laboratory Medicine, 2009, 47, 1311-3.	2.3	7
125	Severe congenital neutropenia: a negative synergistic effect of multiple mutations of ELANE (ELA2) gene. British Journal of Haematology, 2009, 146, 578-580.	2.5	7
126	A novel two bases deletion in the albumin gene causes analbuminaemia in a young Turkish man. Clinica Chimica Acta, 2012, 413, 950-951.	1.1	7

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127	A rare 3q13.31 microdeletion including GAP43 and LSAMP genes. Molecular Cytogenetics, 2013, 6, 52.	0.9	7
128	Are the nail-patella syndrome and the autosomal Goltz-like syndrome the phenotypic expressions of different alleles at the COL5A1 locus?. Human Genetics, 1993, 91, 175-7.	3.8	6
129	Exclusion of the candidate genes ACE and Bcl-2 for six families with nephronophthisis not linked to the NPH1 locus. Nephrology Dialysis Transplantation, 1999, 14, 2328-2331.	0.7	6
130	N-(4-hydroxyphenyl)retinamide inhibitscystogenesis by polycystic epithelial cell lines in vitro. Life Sciences, 1999, 64, PL259-PL265.	4.3	6
131	Stop codon at arginine 586 is the prevalent nephronopthisis type 1 mutation in Italy. Nephrology Dialysis Transplantation, 2006, 21, 2301-2303.	0.7	6
132	A two-base-pairs deletion in the albumin gene causes a new case of analbuminemia. Clinical Chemistry and Laboratory Medicine, 2012, 50, 2221-2223.	2.3	6
133	A novel splicing mutation in the albumin gene (c.270+1G>T) causes analbuminaemia in a German infant. Annals of Clinical Biochemistry, 2016, 53, 615-619.	1.6	6
134	Congenital Analbuminemia in Unrelated Algerian and Turkish Families is Caused by the Same Molecular Defect in the Albumin Gene. Annals of Laboratory Medicine, 2018, 38, 185-188.	2.5	6
135	Refractory Minimal Change Disease and Focal Segmental Glomerular Sclerosis Treated With Anakinra. Kidney International Reports, 2022, 7, 121-124.	0.8	6
136	Towards the identification of (a) gene(s) for autosomal dominant medullary cystic kidney disease. Journal of Nephrology, 2003, 16, 321-8.	2.0	6
137	Case Report: Atypical Manifestations Associated With FOXP3 Mutations. The â€œFil Rougeâ€•of Treg Between IPEX Features and Other Clinical Entities?. Frontiers in Immunology, 2022, 13, 854749.	4.8	6
138	Teaching molecular genetics: chapter 4â€”positional cloning of genetic disorders. Pediatric Nephrology, 2007, 22, 2023-2029.	1.7	5
139	Recurrent lymphomatoid papulosis associated with nephrotic syndrome. An occurrence of uncertain origin. Pediatric Nephrology, 2009, 24, 189-192.	1.7	5
140	A nucleotide deletion and frame-shift cause analbuminemia in a Turkish family. Biochemia Medica, 2016, 26, 264-271.	2.7	5
141	Randomised controlled trial comparing rituximab to mycophenolate mofetil in children and young adults with steroid-dependent idiopathic nephrotic syndrome: study protocol. BMJ Open, 2021, 11, e052450.	1.9	5
142	A new biallelic DNA polymorphism of the human COL5A1 gene. Human Genetics, 1995, 95, 599-600.	3.8	4
143	Juvenile Nephronophthisis and Related Variants: Clinical Features and Molecular Approach. , 2001, 136, 57-67.		4
144	A novel splicing mutation in the ALB gene causing analbuminaemia in a Portuguese woman. Pathology, 2018, 50, 679-682.	0.6	4

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145	A novel insertion (c.1098dupT) in the albumin gene causes analbuminemia in a consanguineous family. European Journal of Medical Genetics, 2019, 62, 144-148.	1.3	4
146	Expression of Collagen by Renal Fibroblasts Treated With FK 506 In Vitro. Transplantation Proceedings, 1998, 30, 957-958.	0.6	3
147	Renal involvement and StrÃ¸mme syndrome. CKJ: Clinical Kidney Journal, 2021, 14, 439-441.	2.9	3
148	mRNA sequencing of a novel NPHS2 intronic mutation in a child with focal and segmental glomerulosclerosis. Saudi Journal of Kidney Diseases and Transplantation: an Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia, 2014, 25, 854.	0.3	3
149	A novel UMOD gene mutation associated with chronic kidney failure at a young age. Clinical Nephrology, 2019, 92, 151-155.	0.7	3
150	The Case | Cystic renal disease, nephrogenic diabetes insipidus, and polycytemia. Kidney International, 2014, 86, 863-864.	5.2	2
151	Schimke immuno-osseous dysplasia, two new cases with peculiar EEG pattern. Brain and Development, 2020, 42, 408-413.	1.1	2
152	A novel nonsense variation in the albumin gene (c.1309 A>T) causing analbuminaemia. British Journal of Biomedical Science, 2021, 78, 154-157.	1.3	2
153	Albumin Benkovac (c.1175 A > G; p.Glu392Gly): a novel genetic variant of human serum albumin. Translational Research, 2010, 155, 118-119.	5.0	1
154	Research update for articles published in <scp>EJCI</scp> in 2013. European Journal of Clinical Investigation, 2015, 45, 1005-1016.	3.4	1
155	Recurrent Hypoglycemia in a Case of Congenital Analbuminemia. Case Reports in Endocrinology, 2020, 2020, 1-6.	0.4	1
156	Atypical presentation of Dent disease in a patient with interstitial Xp11.22 deletion. Journal of Nephrology, 2021, 34, 2111-2115.	2.0	1
157	SP021COQ6 AND COQ2 MUTATIONS ASSOCIATED WITH STEROID RESISTANT NEPHROTIC SYNDROME. Nephrology Dialysis Transplantation, 2017, 32, iii110-iii113.	0.7	0
158	704: Genetic Heterogeneity of Isolated Vesicoureteral Reflux. Journal of Urology, 2005, 173, 192-192.	0.4	0
159	Renal development and cystic diseases. CKJ: Clinical Kidney Journal, 2011, 4, 4.s2.27-4.s2.27.	2.9	0