# Amelie van der Ven

List of Publications by Year
in descending order



| 162 | 12,976 | 22153 | 26613 |
|---|---|---|---|
| papers | citations | 59 h-index | 107 g-index |

| 166 | 166 | 166 | 13429 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Ciliopathies. New England Journal of Medicine, 2011, 364, 1533-1543. | 27.0 | 1,227 |
| 2 | The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. Nature Genetics, 2006, 38, 674-681. | 21.4 | 535 |
| 3 | A Single-Gene Cause in 29.5% of Cases of Steroid-Resistant Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2015, 26, 1279-1289. | 6.1 | 499 |
| 4 | A Dynamic Protein Interaction Landscape of the Human Centrosome-Cilium Interface. Cell, 2015, 163, 1484-1499. | 28.9 | 446 |
| 5 | Patients with Mutations in NPHS2 (Podocin) Do Not Respond to Standard Steroid Treatment of Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2004, 15, 722-732. | 6.1 | 370 |
| 6 | Nephronophthisis-Associated Ciliopathies. Journal of the American Society of Nephrology: JASN, 2007, 18, 1855-1871. | 6.1 | 354 |
| 7 | COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. Journal of Clinical Investigation, 2011, 121, 2013-2024. | 8.2 | 343 |
| 8 | Nephronophthisis. Journal of the American Society of Nephrology: JASN, 2009, 20, 23-35. | 6.1 | 332 |
| 9 | HomozygosityMapper--an interactive approach to homozygosity mapping. Nucleic Acids Research, 2009, 37, W593-W599. | 14.5 | 331 |
| 10 | Ciliopathies. Cold Spring Harbor Perspectives in Biology, 2017, 9, a028191. | 5.5 | 325 |
| 11 | Exploring the genetic basis of early-onset chronic kidney disease. Nature Reviews Nephrology, 2016, 12, 133-146. | 9.6 | 276 |
| 12 | Podocytopathies. Nature Reviews Disease Primers, 2020, 6, 68. | 30.5 | 237 |
| 13 | Mutations in 12 known dominant disease-causing genes clarify many congenital anomalies of the kidney and urinary tract. Kidney International, 2014, 85, 1429-1433. | 5.2 | 203 |
| 14 | Identification of 99 novel mutations in a worldwide cohort of 1,056 patients with a nephronophthisis-related ciliopathy. Human Genetics, 2013, 132, 865-884. | 3.8 | 199 |
| 15 | Single-gene causes of congenital anomalies of the kidney and urinary tract (CAKUT) in humans. Pediatric Nephrology, 2014, 29, 695-704. | 1.7 | 178 |
| 16 | Monogenic causes of chronic kidney disease in adults. Kidney International, 2019, 95, 914-928. | 5.2 | 174 |
| 17 | Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2018, 13, 53-62. | 4.5 | 170 |
| 18 | Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538. | 21.4 | 164 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Fourteen Monogenic Genes Account for 15% of Nephrolithiasis/Nephrocalcinosis. Journal of the American Society of Nephrology: JASN, 2015, 26, 543-551. | 6.1 | 163 |
| 20 | Genetic testing in steroid-resistant nephrotic syndrome: when and how?. Nephrology Dialysis Transplantation, 2016, 31, 1802-1813. | 0.7 | 159 |
| 21 | Immunosuppression and Renal Outcome in Congenital and Pediatric Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 2075-2084. | 4.5 | 153 |
| 22 | Mutations in nuclear pore genes NUP93, NUP205 and XPO5 cause steroid-resistant nephrotic syndrome. Nature Genetics, 2016, 48, 457-465. | 21.4 | 149 |
| 23 | Mutations in DZIP1L, which encodes a ciliary-transition-zone protein, cause autosomal recessive polycystic kidney disease. Nature Genetics, 2017, 49, 1025-1034. | 21.4 | 148 |
| 24 | Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2018, 29, 2348-2361. | 6.1 | 147 |
| 25 | A Systematic Approach to Mapping Recessive Disease Genes in Individuals from Outbred Populations. PLoS Genetics, 2009, 5, e1000353. | 3.5 | 144 |
| 26 | The copy number variation landscape of congenital anomalies of the kidney and urinary tract. Nature Genetics, 2019, 51, 117-127. | 21.4 | 144 |
| 27 | Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis andÂnephrocalcinosis. Kidney International, 2018, 93, 204-213. | 5.2 | 133 |
| 28 | Mutations in FN1 cause glomerulopathy with fibronectin deposits. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 2538-2543. | 7.1 | 125 |
| 29 | The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. Nature Genetics, 2016, 48, 648-656. | 21.4 | 119 |
| 30 | Whole-Exome Sequencing Enables a Precision Medicine Approach for Kidney Transplant Recipients. Journal of the American Society of Nephrology: JASN, 2019, 30, 201-215. | 6.1 | 110 |
| 31 | Novel Insights into the Pathogenesis of Monogenic Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2018, 29, 36-50. | 6.1 | 108 |
| 32 | Whole-exome resequencing reveals recessive mutations in TRAP1 in individuals with CAKUT and VACTERL association. Kidney International, 2014, 85, 1310-1317. | 5.2 | 106 |
| 33 | Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. Clinical Journal of the American Society of Nephrology: CJASN, 2016, 11, 664-672. | 4.5 | 105 |
| 34 | Clinical Features and Histology of Apolipoprotein L1-Associated Nephropathy in the FSGS Clinical Trial. Journal of the American Society of Nephrology: JASN, 2015, 26, 1443-1448. | 6.1 | 104 |
| 35 | Mutations in EMP2 Cause Childhood-Onset Nephrotic Syndrome. American Journal of Human Genetics, 2014, 94, 884-890. | 6.2 | 101 |
| 36 | Genotypeâ€“phenotype correlation in 440 patients with NPHP-related ciliopathies. Kidney International, 2011, 80, 1239-1245. | 5.2 | 99 |

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|---|---|---|---|
| 37 | FAT1 mutations cause a glomerulotubular nephropathy. Nature Communications, 2016, 7, 10822. | 12.8 | 99 |
| 38 | DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. American Journal of Human Genetics, 2015, 96, 81-92. | 6.2 | 98 |
| 39 | Mild Recessive Mutations in Six Fraser Syndromeâ€“Related Genes Cause Isolated Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2014, 25, 1917-1922. | 6.1 | 97 |
| 40 | Exome Sequencing Reveals Cubilin Mutation as a Single-Gene Cause of Proteinuria. Journal of the American Society of Nephrology: JASN, 2011, 22, 1815-1820. | 6.1 | 90 |
| 41 | Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. American Journal of Human Genetics, 2014, 94, 905-914. | 6.2 | 90 |
| 42 | Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. Nature Communications, 2018, 9, 1960. | 12.8 | 90 |
| 43 | Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328. | 8.2 | 89 |
| 44 | Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, . | 8.2 | 89 |
| 45 | Defects of CRB2 Cause Steroid-Resistant Nephrotic Syndrome. American Journal of Human Genetics, 2015, 96, 153-161. | 6.2 | 88 |
| 46 | Genotype/Phenotype Correlation in Nephrotic Syndrome Caused by WT1 Mutations. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 1655-1662. | 4.5 | 87 |
| 47 | Spectrum of mutations in Chinese children with steroid-resistant nephrotic syndrome. Pediatric Nephrology, 2017, 32, 1181-1192. | 1.7 | 81 |
| 48 | A Multi-layered Quantitative InÂVivo Expression Atlas of the Podocyte Unravels Kidney Disease Candidate Genes. Cell Reports, 2018, 23, 2495-2508. | 6.4 | 81 |
| 49 | Exome Sequencing Discerns Syndromes in Patients from Consanguineous Families with Congenital Anomalies of the Kidneys and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2017, 28, 69-75. | 6.1 | 79 |
| 50 | Mutation analysis in nephronophthisis using a combined approach of homozygosity mapping, CEL I endonuclease cleavage, and direct sequencing. Human Mutation, 2008, 29, 418-426. | 2.5 | 76 |
| 51 | Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86. | 6.2 | 75 |
| 52 | Thirteen novel NPHS1 mutations in a large cohort of children with congenital nephrotic syndrome. Nephrology Dialysis Transplantation, 2008, 23, 3527-3533. | 0.7 | 74 |
| 53 | Rapid Detection of Monogenic Causes of Childhood-Onset Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 1109-1116. | 4.5 | 74 |
| 54 | Whole exome sequencing identifies causative mutations in the majority of consanguineous or familial cases with childhood-onset increased renal echogenicity. Kidney International, 2016, 89, 468-475. | 5.2 | 74 |

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|---|---|---|---|
| 55 | Nephronophthisis-Associated CEP164 Regulates Cell Cycle Progression, Apoptosis and Epithelial-to-Mesenchymal Transition. PLoS Genetics, 2014, 10, e1004594. | 3.5 | 73 |
| 56 | Mutations in TBX18 Cause Dominant Urinary Tract Malformations via Transcriptional Dysregulation of Ureter Development. American Journal of Human Genetics, 2015, 97, 291-301. | 6.2 | 72 |
| 57 | Modeling Monogenic Human Nephrotic Syndrome in the Drosophila Garland Cell Nephrocyte. Journal of the American Society of Nephrology: JASN, 2017, 28, 1521-1533. | 6.1 | 70 |
| 58 | Nineteen novel NPHS1 mutations in a worldwide cohort of patients with congenital nephrotic syndrome (CNS). Nephrology Dialysis Transplantation, 2010, 25, 2970-2976. | 0.7 | 69 |
| 59 | Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887. | 5.2 | 67 |
| 60 | Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome. Nature Communications, 2019, 10, 3967. | 12.8 | 66 |
| 61 | Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249. | 1.2 | 66 |
| 62 | Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602. | 6.0 | 64 |
| 63 | Renal-Retinal Ciliopathy Gene Sdccag8 Regulates DNA Damage Response Signaling. Journal of the American Society of Nephrology: JASN, 2014, 25, 2573-2583. | 6.1 | 63 |
| 64 | Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802. | 6.2 | 63 |
| 65 | Recessive nephrocerebellar syndrome on the Galloway-Mowat syndrome spectrum is caused by homozygous protein-truncating mutations ofWDR73. Brain, 2015, 138, 2173-2190. | 7.6 | 60 |
| 66 | Personalized medicine in chronic kidney disease by detection of monogenic mutations. Nephrology Dialysis Transplantation, 2020, 35, 390-397. | 0.7 | 58 |
| 67 | Mutations in WDR4 as a new cause of Gallowayâ€“Mowat syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2460-2465. | 1.2 | 56 |
| 68 | Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316. | 6.2 | 56 |
| 69 | SDCCAG8 Regulates Pericentriolar Material Recruitment and Neuronal Migration in the Developing Cortex. Neuron, 2014, 83, 805-822. | 8.1 | 52 |
| 70 | Secreted metalloproteases ADAMTS9 and ADAMTS20 have a non-canonical role in ciliary vesicle growth during ciliogenesis. Nature Communications, 2019, 10, 953. | 12.8 | 51 |
| 71 | MKS1 regulates ciliary INPP5E levels in Joubert syndrome. Journal of Medical Genetics, 2016, 53, 62-72. | 3.2 | 48 |
| 72 | Rare Variants in BNC2 Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. American Journal of Human Genetics, 2019, 104, 994-1006. | 6.2 | 47 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Targeted Resequencing of 29 Candidate Genes and Mouse Expression Studies ImplicateZIC3andFOXF1in Human VATER/VACTERL Association. Human Mutation, 2015, 36, 1150-1154. | 2.5 | 46 |
| 74 | Mutations in COQ8B (ADCK4) found in patients with steroid-resistant nephrotic syndrome alter COQ8B function. Human Mutation, 2018, 39, 406-414. | 2.5 | 43 |
| 75 | WDR73Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. Human Mutation, 2015, 36, 1021-1028. | 2.5 | 42 |
| 76 | GAPVD1 and ANKFY1 Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2018, 29, 2123-2138. | 6.1 | 42 |
| 77 | Whole exome sequencing identified ATP6V1C2 as a novel candidate gene for recessive distal renal tubular acidosis. Kidney International, 2020, 97, 567-579. | 5.2 | 42 |
| 78 | Exome sequencing in Jewish and Arab patients with rhabdomyolysis reveals single-gene etiology in 43% of cases. Pediatric Nephrology, 2017, 32, 2273-2282. | 1.7 | 40 |
| 79 | Multisystem inflammation and susceptibility to viral infections in human ZNFX1 deficiency. Journal of Allergy and Clinical Immunology, 2021, 148, 381-393. | 2.9 | 40 |
| 80 | Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. Journal of Medical Genetics, 2016, 53, 208-214. | 3.2 | 39 |
| 81 | Advillin acts upstream of phospholipase C Ïµ1 in steroid-resistant nephrotic syndrome. Journal of Clinical Investigation, 2017, 127, 4257-4269. | 8.2 | 39 |
| 82 | ADCK4 Deficiency Destabilizes the Coenzyme Q Complex, Which Is Rescued by 2,4-Dihydroxybenzoic Acid Treatment. Journal of the American Society of Nephrology: JASN, 2020, 31, 1191-1211. | 6.1 | 38 |
| 83 | ZMYND10 stabilizes intermediate chain proteins in the cytoplasmic pre-assembly of dynein arms. PLoS Genetics, 2018, 14, e1007316. | 3.5 | 37 |
| 84 | Treatment with 2,4-Dihydroxybenzoic Acid Prevents FSGS Progression and Renal Fibrosis in Podocyte-Specific Coq6 Knockout Mice. Journal of the American Society of Nephrology: JASN, 2019, 30, 393-405. | 6.1 | 36 |
| 85 | Whole exome sequencing in childhood-onset lupus frequently detects single gene etiologies. Pediatric Rheumatology, 2019, 17, 52. | 2.1 | 34 |
| 86 | Mutation of Growth Arrest Specific 8 Reveals a Role in Motile Cilia Function and Human Disease. PLoS Genetics, 2016, 12, e1006220. | 3.5 | 33 |
| 87 | IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665. | 3.2 | 32 |
| 88 | Clinical, biochemical, and pathophysiological analysis of SLC34A1 mutations. Physiological Reports, 2018, 6, e13715. | 1.7 | 32 |
| 89 | Dominant PAX2 mutations may cause steroid-resistant nephrotic syndrome and FSGS in children. Pediatric Nephrology, 2019, 34, 1607-1613. | 1.7 | 31 |
| 90 | Analysis of 24 genes reveals a monogenic cause in 11.1% of cases with steroid-resistant nephrotic syndrome at a single center. Pediatric Nephrology, 2018, 33, 305-314. | 1.7 | 30 |

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| 91 | Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. American Journal of Human Genetics, 2017, 100, 323-333. | 6.2 | 29 |
| 92 | Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. American Journal of Human Genetics, 2019, 104, 45-54. | 6.2 | 29 |
| 93 | Homozygous frameshift mutations in FAT1 cause a syndrome characterized by colobomatous-microphthalmia, ptosis, nephropathy and syndactyly. Nature Communications, 2019, 10, 1180. | 12.8 | 27 |
| 94 | Gene panel sequencing identifies a likely monogenic cause in 7% of 235 Pakistani families with nephrolithiasis. Human Genetics, 2019, 138, 211-219. | 3.8 | 26 |
| 95 | TBC1D8B Mutations Implicate RAB11-Dependent Vesicular Trafficking in the Pathogenesis of Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2019, 30, 2338-2353. | 6.1 | 25 |
| 96 | Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742. | 6.2 | 25 |
| 97 | A FANCD2/FANCI-Associated Nuclease 1-Knockout Model Develops Karyomegalic Interstitial Nephritis. Journal of the American Society of Nephrology: JASN, 2016, 27, 3552-3559. | 6.1 | 24 |
| 98 | PLCE1 regulates the migration, proliferation, and differentiation of podocytes. Experimental and Molecular Medicine, 2020, 52, 594-603. | 7.7 | 24 |
| 99 | Mutations in KIRREL1, a slit diaphragm component, cause steroid-resistant nephrotic syndrome. Kidney International, 2019, 96, 883-889. | 5.2 | 23 |
| 100 | Whole Exome Sequencing Reveals a Monogenic Cause of Disease in â‰ˆ43% of 35 Families With Midaortic Syndrome. Hypertension, 2018, 71, 691-699. | 2.7 | 22 |
| 101 | Human urine-derived renal epithelial cells provide insights into kidney-specific alternate splicing variants. European Journal of Human Genetics, 2018, 26, 1791-1796. | 2.8 | 22 |
| 102 | Genetic variants in the LAMA5 gene in pediatric nephrotic syndrome. Nephrology Dialysis Transplantation, 2019, 34, 485-493. | 0.7 | 22 |
| 103 | Mouse genetics reveals Barttin as a genetic modifier of Joubert syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 1113-1118. | 7.1 | 22 |
| 104 | Responsiveness of sphingosine phosphate lyase insufficiency syndrome to vitamin <scp>B6</scp> cofactor supplementation. Journal of Inherited Metabolic Disease, 2020, 43, 1131-1142. | 3.6 | 21 |
| 105 | Recessive NOS1AP variants impair actin remodeling and cause glomerulopathy in humans and mice. Science Advances, 2021, 7, . | 10.3 | 21 |
| 106 | Osteoclast stimulation factor 1 (Ostf1) KNOCKOUT increases trabecular bone mass in mice. Mammalian Genome, 2017, 28, 498-514. | 2.2 | 19 |
| 107 | SDCCAG8 Interacts with RAB Effector Proteins RABEP2 and ERC1 and Is Required for Hedgehog Signaling. PLoS ONE, 2016, 11, e0156081. | 2.5 | 19 |
| 108 | Acute multi-sgRNA knockdown of KEOPS complex genes reproduces the microcephaly phenotype of the stable knockout zebrafish model. PLoS ONE, 2018, 13, e0191503. | 2.5 | 18 |

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|---|---|---|---|
| 109 | CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. American Journal of Human Genetics, 2019, 105, 1286-1293. | 6.2 | 18 |
| 110 | Copy Number Variant Analysis and Genome-wide Association Study Identify Loci with Large Effect for Vesicoureteral Reflux. Journal of the American Society of Nephrology: JASN, 2021, 32, 805-820. | 6.1 | 17 |
| 111 | Galloway-Mowat syndrome in Taiwan: OSGEP mutation and unique clinical phenotype. Orphanet Journal of Rare Diseases, 2018, 13, 226. | 2.7 | 16 |
| 112 | Effects of Diet and Social Housing on Reproductive Success in Adult Zebrafish, Danio rerio. Zebrafish, 2018, 15, 445-453. | 1.1 | 16 |
| 113 | Novel homozygous ENPP1 mutation causes generalized arterial calcifications of infancy, thrombocytopenia, and cardiovascular and central nervous system syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2112-2118. | 1.2 | 16 |
| 114 | Whole-Exome Sequencing in Critically Ill Neonates and Infants: Diagnostic Yield and Predictability of Monogenic Diagnosis. Neonatology, 2021, 118, 454-461. | 2.0 | 16 |
| 115 | Deep learning is widely applicable to phenotyping embryonic development and disease. Development (Cambridge), 2021, 148, . | 2.5 | 16 |
| 116 | Targeted sequencing of 96 renal developmental microRNAs in 1213 individuals from 980 families with congenital anomalies of the kidney and urinary tract. Nephrology Dialysis Transplantation, 2016, 31, 1280-1283. | 0.7 | 15 |
| 117 | HSPA6: A new autosomal recessive candidate gene for the VATER/VACTERL malformation spectrum. Birth Defects Research, 2019, 111, 591-597. | 1.5 | 15 |
| 118 | Utility of Genomic Testing after Renal Biopsy. American Journal of Nephrology, 2020, 51, 43-53. | 3.1 | 15 |
| 119 | Mutations in PRDM15 Are a Novel Cause of Galloway-Mowat Syndrome. Journal of the American Society of Nephrology: JASN, 2021, 32, 580-596. | 6.1 | 15 |
| 120 | Inhibition of endoplasmic reticulum stress signaling rescues cytotoxicity of human apolipoprotein-L1 risk variants in Drosophila. Kidney International, 2022, 101, 1216-1231. | 5.2 | 15 |
| 121 | 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 |
| 122 | The utility of a genetic kidney disease clinic employing a broad range of genomic testing platforms: experience of the Irish Kidney Gene Project. Journal of Nephrology, 2022, 35, 1655-1665. | 2.0 | 14 |
| 123 | The nucleoside-diphosphate kinase NME3 associates with nephronophthisis proteins and is required for ciliary function during renal development. Journal of Biological Chemistry, 2018, 293, 15243-15255. | 3.4 | 13 |
| 124 | Panel sequencing distinguishes monogenic forms of nephritis from nephrosis in children. Nephrology Dialysis Transplantation, 2019, 34, 474-485. | 0.7 | 13 |
| 125 | COL4A1 mutations as a potential novel cause of autosomal dominant CAKUT in humans. Human Genetics, 2019, 138, 1105-1115. | 3.8 | 13 |
| 126 | Disruption of MAGI2-RapGEF2-Rap1 signaling contributes to podocyte dysfunction in congenital nephrotic syndrome caused by mutations in MAGI2. Kidney International, 2019, 96, 642-655. | 5.2 | 13 |

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| 127 | Beyond the tubule: pathological variants of LRP2, encoding the megalin receptor, result in glomerular loss and early progressive chronic kidney disease. American Journal of Physiology - Renal Physiology, 2020, 319, F988-F999. | 2.7 | 13 |
| 128 | PRDM15 loss of function links NOTCH and WNT/PCP signaling to patterning defects in holoprosencephaly. Science Advances, 2020, 6, eaax9852. | 10.3 | 13 |
| 129 | A recurrent, homozygous EMC10 frameshift variant is associated with a syndrome of developmental delay with variable seizures and dysmorphic features. Genetics in Medicine, 2021, 23, 1158-1162. | 2.4 | 13 |
| 130 | Cystin genetic variants cause autosomal recessive polycystic kidney disease associated with altered Myc expression. Scientific Reports, 2021, 11, 18274. | 3.3 | 13 |
| 131 | Reverse phenotyping facilitates disease allele calling in exome sequencing of patients with CAKUT. Genetics in Medicine, 2022, 24, 307-318. | 2.4 | 13 |
| 132 | Genetic dissection of kidney disorders. Nature Reviews Nephrology, 2015, 11, 635-636. | 9.6 | 12 |
| 133 | Roscovitine blocks collecting duct cyst growth in Cep164-deficient kidneys. Kidney International, 2019, 96, 320-326. | 5.2 | 12 |
| 134 | DAAM2 Variants Cause Nephrotic Syndrome via Actin Dysregulation. American Journal of Human Genetics, 2020, 107, 1113-1128. | 6.2 | 12 |
| 135 | A discarded synonymous variant in NPHP3 explains nephronophthisis and congenital hepatic fibrosis in several families. Human Mutation, 2021, 42, 1221-1228. | 2.5 | 12 |
| 136 | Cystic kidneys in fetal Walkerâ€“Warburg syndrome with POMT2 mutation: Intrafamilial phenotypic variability in four siblings and review of literature. American Journal of Medical Genetics, Part A, 2017, 173, 2697-2702. | 1.2 | 11 |
| 137 | Progressive Pseudorheumatoid Dysplasia resolved by whole exome sequencing: a novel mutation in WISP3 and review of the literature. BMC Medical Genetics, 2019, 20, 53. | 2.1 | 11 |
| 138 | Loss ofAnks6leads to YAP deficiency and liver abnormalities. Human Molecular Genetics, 2020, 29, 3064-3080. | 2.9 | 11 |
| 139 | Immunological Impact of a Gluten-Free Dairy-Free Diet in Children With Kidney Disease: A Feasibility Study. Frontiers in Immunology, 2021, 12, 624821. | 4.8 | 11 |
| 140 | A small molecule screening to detect potential therapeutic targets in human podocytes. American Journal of Physiology - Renal Physiology, 2017, 312, F157-F171. | 2.7 | 10 |
| 141 | Corticosteroid treatment exacerbates nephrotic syndrome in a zebrafish model of magi2a knockout. Kidney International, 2019, 95, 1079-1090. | 5.2 | 9 |
| 142 | ANK3 related neurodevelopmental disorders: expanding the spectrum of heterozygous loss-of-function variants. Neurogenetics, 2021, 22, 263-269. | 1.4 | 8 |
| 143 | Whole-Exome Sequencing Reveals FAT4 Mutations in a Clinically Unrecognizable Patient with Syndromic CAKUT: A Case Report. Molecular Syndromology, 2017, 8, 272-277. | 0.8 | 7 |
| 144 | Recessive Mutations in SYNPO2 as a Candidate of Monogenic Nephrotic Syndrome. Kidney International Reports, 2021, 6, 472-483. | 0.8 | 7 |

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|---|---|---|---|
| 145 | Novel nephronophthisis-associated variants reveal functional importance of MAPKBP1 dimerization for centriolar recruitment. Kidney International, 2020, 98, 958-969. | 5.2 | 6 |
| 146 | Exome survey of individuals affected by VATER / VACTERL with renal phenotypes identifies phenocopies and novel candidate genes. American Journal of Medical Genetics, Part A, 2021, 185, 3784-3792. | 1.2 | 6 |
| 147 | Whole-exome sequencing identifiesFOXL2,FOXA2andFOXA3as candidate genes for monogenic congenital anomalies of the kidneys and urinary tract. Nephrology Dialysis Transplantation, 2022, 37, 1833-1843. | 0.7 | 6 |
| 148 | Ttc30a affects tubulin modifications in a model for ciliary chondrodysplasia with polycystic kidney disease. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, . | 7.1 | 6 |
| 149 | Proteomic analysis identifies ZMYM2 as endogenous binding partner of TBX18 protein in 293 and A549 cells. Biochemical Journal, 2022, 479, 91-109. | 3.7 | 6 |
| 150 | Expression of a Truncated Form of ODAD1 Associated with an Unusually Mild Primary Ciliary Dyskinesia Phenotype. International Journal of Molecular Sciences, 2022, 23, 1753. | 4.1 | 6 |
| 151 | Homozygous <scp>WNT9B</scp> variants in two families with bilateral renal agenesis/hypoplasia/dysplasia. American Journal of Medical Genetics, Part A, 2021, 185, 3005-3011. | 1.2 | 5 |
| 152 | Prevalence and clinical prediction of mitochondrial disorders in a large neuropediatric cohort. Clinical Genetics, 2021, 100, 766-770. | 2.0 | 5 |
| 153 | A homozygous missense variant in VWA2, encoding an interactor of the Fraser-complex, in a patient with vesicoureteral reflux. PLoS ONE, 2018, 13, e0191224. | 2.5 | 5 |
| 154 | Karyomegalic interstitial nephritis. Lancet, The, 2013, 382, 2093. | 13.7 | 2 |
| 155 | Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. Kidney International Reports, 2021, 6, 460-471. | 0.8 | 2 |
| 156 | A truncating NRIP1 variant in an Arabic family with congenital anomalies of the kidneys and urinary tract. American Journal of Medical Genetics, Part A, 2021, , . | 1.2 | 2 |
| 157 | Whole exome sequencing identifies potential candidate genes for spina bifida derived from mouse models. American Journal of Medical Genetics, Part A, 2022, , . | 1.2 | 2 |
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