

# Daniela Anne Braun

## List of Publications by Year in descending order

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Version: 2024-02-01

50  
papers

4,269  
citations

126907

33  
h-index

175258

52  
g-index

53  
all docs

53  
docs citations

53  
times ranked

6234  
citing authors

#	ARTICLE	IF	CITATIONS
1	Ciliopathies. Cold Spring Harbor Perspectives in Biology, 2017, 9, a028191.	5.5	325
2	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
3	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	6.2	196
4	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 93, 672-686.	6.2	184
5	Monogenic causes of chronic kidney disease in adults. Kidney International, 2019, 95, 914-928.	5.2	174
6	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
7	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	21.4	164
8	Fourteen Monogenic Genes Account for 15% of Nephrolithiasis/Nephrocalcinosis. Journal of the American Society of Nephrology: JASN, 2015, 26, 543-551.	6.1	163
9	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. Journal of Clinical Investigation, 2017, 127, 912-928.	8.2	160
10	Mutations in nuclear pore genes NUP93, NUP205 and XPO5 cause steroid-resistant nephrotic syndrome. Nature Genetics, 2016, 48, 457-465.	21.4	149
11	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
12	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. American Journal of Human Genetics, 2013, 93, 711-720.	6.2	135
13	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis and Nephrocalcinosis. Kidney International, 2018, 93, 204-213.	5.2	133
14	The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. Nature Genetics, 2016, 48, 648-656.	21.4	119
15	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
16	High-throughput mutation analysis in patients with a nephronophthisis-associated ciliopathy applying multiplexed barcoded array-based PCR amplification and next-generation sequencing. Journal of Medical Genetics, 2012, 49, 756-767.	3.2	109
17	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
18	Decreased number of circulating progenitor cells in obesity: beneficial effects of weight reduction. European Heart Journal, 2008, 29, 1560-1568.	2.2	104

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19	FAT1 mutations cause a glomerulotubular nephropathy. <i>Nature Communications</i> , 2016, 7, 10822.	12.8	99
20	DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 96, 81-92.	6.2	98
21	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 905-914.	6.2	90
22	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. <i>Nature Communications</i> , 2018, 9, 1960.	12.8	90
23	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2018, 128, 4313-4328.	8.2	89
24	A Multi-layered Quantitative In Vivo Expression Atlas of the Podocyte Unravels Kidney Disease Candidate Genes. <i>Cell Reports</i> , 2018, 23, 2495-2508.	6.4	81
25	Whole exome sequencing identifies causative mutations in the majority of consanguineous or familial cases with childhood-onset increased renal echogenicity. <i>Kidney International</i> , 2016, 89, 468-475.	5.2	74
26	Modeling Monogenic Human Nephrotic Syndrome in the Drosophila Garland Cell Nephrocyte. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 1521-1533.	6.1	70
27	Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome. <i>Nature Communications</i> , 2019, 10, 3967.	12.8	66
28	Mutations in <i>WDR4</i> as a new cause of Galloway-Mowat syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2460-2465.	1.2	56
29	The Vac14-interaction Network Is Linked to Regulators of the Endolysosomal and Autophagic Pathway. <i>Molecular and Cellular Proteomics</i> , 2014, 13, 1397-1411.	3.8	51
30	Targeted Resequencing of 29 Candidate Genes and Mouse Expression Studies Implicate <i>ZIC3</i> and <i>FOXF1</i> in Human VATER/VACTERL Association. <i>Human Mutation</i> , 2015, 36, 1150-1154.	2.5	46
31	GAPVD1 and ANKFY1 Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2123-2138.	6.1	42
32	Mutations in SLC26A1 Cause Nephrolithiasis. <i>American Journal of Human Genetics</i> , 2016, 98, 1228-1234.	6.2	41
33	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. <i>Journal of Medical Genetics</i> , 2016, 53, 208-214.	3.2	39
34	Advillin acts upstream of phospholipase C $\mu$ 1 in steroid-resistant nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2017, 127, 4257-4269.	8.2	39
35	<i>IFT81</i> , encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. <i>Journal of Medical Genetics</i> , 2015, 52, 657-665.	3.2	32
36	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. <i>American Journal of Human Genetics</i> , 2017, 100, 323-333.	6.2	29

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37	Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. American Journal of Human Genetics, 2019, 104, 45-54.	6.2	29
38	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
39	Whole Exome Sequencing Reveals a Monogenic Cause of Disease in 43% of 35 Families With Midaortic Syndrome. Hypertension, 2018, 71, 691-699.	2.7	22
40	Genetic variants in the LAMA5 gene in pediatric nephrotic syndrome. Nephrology Dialysis Transplantation, 2019, 34, 485-493.	0.7	22
41	Recessive <i>NOS1AP</i> variants impair actin remodeling and cause glomerulopathy in humans and mice. Science Advances, 2021, 7, .	10.3	21
42	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
43	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
44	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
45	Panel sequencing distinguishes monogenic forms of nephritis from nephrosis in children. Nephrology Dialysis Transplantation, 2019, 34, 474-485.	0.7	13
46	Cystin genetic variants cause autosomal recessive polycystic kidney disease associated with altered Myc expression. Scientific Reports, 2021, 11, 18274.	3.3	13
47	DAAM2 Variants Cause Nephrotic Syndrome via Actin Dysregulation. American Journal of Human Genetics, 2020, 107, 1113-1128.	6.2	12
48	Cystic kidneys in fetal Walker-Warburg syndrome with <i>POMT2</i> 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
49	Novel nephronophthisis-associated variants reveal functional importance of MAPKBP1 dimerization for centriolar recruitment. Kidney International, 2020, 98, 958-969.	5.2	6
50	Gene mutation analysis in Iranian children with nephronophthisis: a two-center study. Iranian Journal of Kidney Diseases, 2015, 9, 119-25.	0.1	2