

# Jan Halbritter

## List of Publications by Year in descending order

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61  
papers

3,741  
citations

172207

29  
h-index

155451

55  
g-index

63  
all docs

63  
docs citations

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times ranked

5329  
citing authors

#	ARTICLE	IF	CITATIONS
1	A Single-Gene Cause in 29.5% of Cases of Steroid-Resistant Nephrotic Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 1279-1289.	3.0	499
2	Identification of 99 novel mutations in a worldwide cohort of 1,056 patients with a nephronophthisis-related ciliopathy. <i>Human Genetics</i> , 2013, 132, 865-884.	1.8	199
3	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. <i>American Journal of Human Genetics</i> , 2013, 93, 915-925.	2.6	196
4	Mutations in <i>RSPH1</i> Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2014, 189, 707-717.	2.5	191
5	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies <i>C21orf59</i> and <i>CCDC65</i> Defects as Causing Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2013, 93, 672-686.	2.6	184
6	<i>ANKS6</i> is a central component of a nephronophthisis module linking <i>NEK8</i> to <i>INVS</i> and <i>NPHP3</i> . <i>Nature Genetics</i> , 2013, 45, 951-956.	9.4	183
7	<i>ZMYND10</i> Is Mutated in Primary Ciliary Dyskinesia and Interacts with <i>LRR6</i> . <i>American Journal of Human Genetics</i> , 2013, 93, 336-345.	2.6	183
8	Fourteen Monogenic Genes Account for 15% of Nephrolithiasis/Nephrocalcinosis. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 543-551.	3.0	163
9	Mutations in <i>SPAG1</i> Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. <i>American Journal of Human Genetics</i> , 2013, 93, 711-720.	2.6	135
10	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis and nephrocalcinosis. <i>Kidney International</i> , 2018, 93, 204-213.	2.6	133
11	High-throughput mutation analysis in patients with a nephronophthisis-associated ciliopathy applying multiplexed barcoded array-based PCR amplification and next-generation sequencing. <i>Journal of Medical Genetics</i> , 2012, 49, 756-767.	1.5	109
12	Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2016, 11, 664-672.	2.2	105
13	<i>FAT1</i> mutations cause a glomerulotubular nephropathy. <i>Nature Communications</i> , 2016, 7, 10822.	5.8	99
14	<i>DCDC2</i> Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 96, 81-92.	2.6	98
15	<i>TMEM231</i> , mutated in orofacioidigital and Meckel syndromes, organizes the ciliary transition zone. <i>Journal of Cell Biology</i> , 2015, 209, 129-142.	2.3	95
16	Mutations of <i>CEP83</i> Cause Infantile Nephronophthisis and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 905-914.	2.6	90
17	Mutations in <i>CSPP1</i> Lead to Classical Joubert Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 80-86.	2.6	75
18	Rapid Detection of Monogenic Causes of Childhood-Onset Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2014, 9, 1109-1116.	2.2	74

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19	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.	2.6	74
20	Mutation of the Mg <sup>2+</sup> Transporter SLC41A1 Results in a Nephronophthisis-Like Phenotype. <i>Journal of the American Society of Nephrology: JASN</i> , 2013, 24, 967-977.	3.0	63
21	Cystinuria: clinical practice recommendation. <i>Kidney International</i> , 2021, 99, 48-58.	2.6	58
22	Value of renal gene panel diagnostics in adults waiting for kidney transplantation due to undetermined end-stage renal disease. <i>Kidney International</i> , 2019, 96, 222-230.	2.6	47
23	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.	1.1	46
24	Genetics in chronic kidney disease: conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. <i>Kidney International</i> , 2022, 101, 1126-1141.	2.6	46
25	Mutations in SLC26A1 Cause Nephrolithiasis. <i>American Journal of Human Genetics</i> , 2016, 98, 1228-1234.	2.6	41
26	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. <i>Journal of Medical Genetics</i> , 2016, 53, 208-214.	1.5	39
27	Refining genotype-phenotype correlations in 304 patients with autosomal recessive polycystic kidney disease and PKHD1 gene variants. <i>Kidney International</i> , 2021, 100, 650-659.	2.6	38
28	Genetic and Evolutionary Analyses of the Human Bone Morphogenetic Protein Receptor 2 (BMP2) in the Pathophysiology of Obesity. <i>PLoS ONE</i> , 2011, 6, e16155.	1.1	38
29	Role of genetic variation in the human sodium-glucose cotransporter 2 gene ( <i>SGLT2</i> ) in glucose homeostasis. <i>Pharmacogenomics</i> , 2011, 12, 1119-1126.	0.6	37
30	<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.	1.5	32
31	Clinical, biochemical, and pathophysiological analysis of <i>SLC34A1</i> mutations. <i>Physiological Reports</i> , 2018, 6, e13715.	0.7	32
32	Matching clinical and genetic diagnoses in autosomal dominant polycystic kidney disease reveals novel phenocopies and potential candidate genes. <i>Genetics in Medicine</i> , 2020, 22, 1374-1383.	1.1	30
33	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. <i>American Journal of Human Genetics</i> , 2017, 100, 323-333.	2.6	29
34	Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. <i>American Journal of Human Genetics</i> , 2019, 104, 45-54.	2.6	29
35	Gene panel sequencing identifies a likely monogenic cause in 7% of 235 Pakistani families with nephrolithiasis. <i>Human Genetics</i> , 2019, 138, 211-219.	1.8	26
36	Biallelic Expression of Mucin-1 in Autosomal Dominant Tubulointerstitial Kidney Disease: Implications for Nongenetic Disease Recognition. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2298-2309.	3.0	25

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37	A Deregulated Stress Response Underlies Distinct INF2-Associated Disease Profiles. <i>Journal of the American Society of Nephrology: JASN</i> , 2020, 31, 1296-1313.	3.0	20
38	Claudin-10a Deficiency Shifts Proximal Tubular Cl- Permeability to Cation Selectivity via Claudin-2 Redistribution. <i>Journal of the American Society of Nephrology: JASN</i> , 2022, 33, 699-717.	3.0	20
39	Diagnosing FSGS without kidney biopsy – a novel INF2-mutation in a family with ESRD of unknown origin. <i>BMC Medical Genetics</i> , 2016, 17, 73.	2.1	16
40	THOC5: a novel gene involved in HDL-cholesterol metabolism. <i>Journal of Lipid Research</i> , 2013, 54, 3170-3176.	2.0	15
41	Evaluating pathogenicity of SLC34A3-Ser192Leu, a frequent European missense variant in disorders of renal phosphate wasting. <i>Urolithiasis</i> , 2019, 47, 511-519.	1.2	15
42	Update on Hereditary Kidney Stone Disease and Introduction of a New Clinical Patient Registry in Germany. <i>Frontiers in Pediatrics</i> , 2018, 6, 47.	0.9	14
43	Posttransplant nephrotic syndrome resulting from NELL1-positive membranous nephropathy. <i>American Journal of Transplantation</i> , 2021, 21, 3175-3179.	2.6	14
44	The nucleoside-diphosphate kinase NME3 associates with nephronophthisis proteins and is required for ciliary function during renal development. <i>Journal of Biological Chemistry</i> , 2018, 293, 15243-15255.	1.6	13
45	Genetics of kidney stone disease – Polygenic meets monogenic. <i>Nephrologie Et Therapeutique</i> , 2021, 17, S88-S94.	0.2	10
46	Defective claudin-10 causes a novel variation of HELIX syndrome through compromised tight junction strand assembly. <i>Genes and Diseases</i> , 2022, 9, 1301-1314.	1.5	9
47	Acute kidney injury and its progression in hospitalized patients – Results from a retrospective multicentre cohort study with a digital decision support system. <i>PLoS ONE</i> , 2021, 16, e0254608.	1.1	9
48	Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. <i>Kidney International</i> , 2022, 101, 1039-1053.	2.6	8
49	Isolated Renal Relapse of Sarcoidosis under Low-Dose Glucocorticoid Therapy. <i>Journal of General Internal Medicine</i> , 2008, 23, 879-882.	1.3	7
50	Retrospective genetic analysis illustrates the spectrum of autosomal Alport syndrome in a case of living-related donor kidney transplantation. <i>BMC Nephrology</i> , 2019, 20, 340.	0.8	7
51	Novel nephronophthisis-associated variants reveal functional importance of MAPKBP1 dimerization for centriolar recruitment. <i>Kidney International</i> , 2020, 98, 958-969.	2.6	6
52	Effective immunosuppressive management with belatacept and eculizumab in post-transplant aHUS due to a homozygous deletion of CFHR1/CFHR3 and the presence of CFH antibodies. <i>CKJ: Clinical Kidney Journal</i> , 2017, 10, 742-746.	1.4	5
53	Successful Simultaneous Pancreas Kidney Transplantation in Maturity-Onset Diabetes of the Young Type 5. <i>Transplantation</i> , 2011, 92, e45-e47.	0.5	3
54	Deleterious Impact of a Novel CFH Splice Site Variant in Atypical Hemolytic Uremic Syndrome. <i>Frontiers in Genetics</i> , 2019, 10, 465.	1.1	3

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55	Autosomal dominant polycystic kidney disease in absence of renal cyst formation illustrates genetic interaction between WT1 and PKD1. <i>Journal of Medical Genetics</i> , 2021, 58, 140-144.	1.5	2
56	Challenging Disease Ontology by Instances of Atypical PKHD1 and PKD1 Genetics. <i>Frontiers in Genetics</i> , 2021, 12, 682565.	1.1	2
57	Novel somatic <i>PBX1</i> mosaicism likely masking syndromic CAKUT in an adult with bilateral kidney hypoplasia. <i>CKJ: Clinical Kidney Journal</i> , 0, , .	1.4	2
58	Early Flow Disturbances of Tunnelled Haemodialysis Catheters and Topographic Landmarks in Chest X-Ray. <i>Blood Purification</i> , 2018, 46, 70-76.	0.9	0
59	The Case   Atypical cysts in a patient with autosomal dominant polycystic kidney disease. <i>Kidney International</i> , 2019, 96, 1043-1044.	2.6	0
60	â€œCase of the Monthâ€™ from the University Medicine Mannheim: managing a complex stone patient with recurrent stone formation. <i>BJU International</i> , 2021, 127, 402-408.	1.3	0
61	MO047: Biallelic pathogenic variants in <i>ROBO1</i> associate with syndromic CAKUT. <i>Nephrology Dialysis Transplantation</i> , 2022, 37, .	0.4	0