Jan Halbritter

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3055164/publications.pdf

Version: 2024-02-01

61 papers	3,741 citations	172207 29 h-index	155451 55 g-index
63 all docs	63 docs citations	63 times ranked	5329 citing authors

#	Article	IF	Citations
1	Defective claudin-10 causes a novel variation of HELIX syndrome through compromised tight junction strand assembly. Genes and Diseases, 2022, 9, 1301-1314.	1.5	9
2	Claudin-10a Deficiency Shifts Proximal Tubular Cl- Permeability to Cation Selectivity via Claudin-2 Redistribution. Journal of the American Society of Nephrology: JASN, 2022, 33, 699-717.	3.0	20
3	Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. Kidney International, 2022, 101, 1039-1053.	2.6	8
4	Genetics in chronic kidney disease: conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney International, 2022, 101, 1126-1141.	2.6	46
5	MO047: Biallelic pathogenic variants in ROBO1 associate with syndromic CAKUT. Nephrology Dialysis Transplantation, 2022, 37, .	0.4	0
6	Autosomal dominant polycystic kidney disease in absence of renal cyst formation illustrates genetic interaction between WT1 and PKD1. Journal of Medical Genetics, 2021, 58, 140-144.	1.5	2
7	Cystinuria: clinical practice recommendation. Kidney International, 2021, 99, 48-58.	2.6	58
8	â€˜Case of the Monthâ€™ from the University Medicine Mannheim: managing a complex stone patient with recurrent stone formation. BJU International, 2021, 127, 402-408.	1.3	0
9	Genetics of kidney stone diseaseâ€”Polygenic meets monogenic. Nephrologie Et Therapeutique, 2021, 17, S88-S94.	0.2	10
10	Posttransplant nephrotic syndrome resulting from NELL1-positive membranous nephropathy. American Journal of Transplantation, 2021, 21, 3175-3179.	2.6	14
11	Challenging Disease Ontology by Instances of Atypical PKHD1 and PKD1 Genetics. Frontiers in Genetics, 2021, 12, 682565.	1.1	2
12	Acute kidney injury and its progression in hospitalized patientsâ€”Results from a retrospective multicentre cohort study with a digital decision support system. PLoS ONE, 2021, 16, e0254608.	1.1	9
13	Refining genotypeâ€“phenotype correlations in 304 patients with autosomal recessive polycystic kidney disease and PKHD1 gene variants. Kidney International, 2021, 100, 650-659.	2.6	38
14	Matching clinical and genetic diagnoses in autosomal dominant polycystic kidney disease reveals novel phenocopies and potential candidate genes. Genetics in Medicine, 2020, 22, 1374-1383.	1.1	30
15	Novel nephronophthisis-associated variants reveal functional importance of MAPKBP1 dimerization for centriolar recruitment. Kidney International, 2020, 98, 958-969.	2.6	6
16	A Deregulated Stress Response Underlies Distinct INF2-Associated Disease Profiles. Journal of the American Society of Nephrology: JASN, 2020, 31, 1296-1313.	3.0	20
17	Retrospective genetic analysis illustrates the spectrum of autosomal Alport syndrome in a case of living-related donor kidney transplantation. BMC Nephrology, 2019, 20, 340.	0.8	7
18	The Case | Atypical cysts in a patient with autosomal dominant polycystic kidney disease. Kidney International, 2019, 96, 1043-1044.	2.6	0

#	Article	IF	Citations
19	Deleterious Impact of a Novel CFH Splice Site Variant in Atypical Hemolytic Uremic Syndrome. Frontiers in Genetics, 2019, 10, 465.	1.1	3
20	Value of renal gene panel diagnostics in adults waiting for kidney transplantation due to undetermined end-stage renal disease. Kidney International, 2019, 96, 222-230.	2.6	47
21	Evaluating pathogenicity of SLC34A3-Ser192Leu, a frequent European missense variant in disorders of renal phosphate wasting. Urolithiasis, 2019, 47, 511-519.	1.2	15
22	Gene panel sequencing identifies a likely monogenic cause in 7% of 235 Pakistani families with nephrolithiasis. Human Genetics, 2019, 138, 211-219.	1.8	26
23	Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. American Journal of Human Genetics, 2019, 104, 45-54.	2.6	29
24	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis andÂnephrocalcinosis. Kidney International, 2018, 93, 204-213.	2.6	133
25	Clinical, biochemical, and pathophysiological analysis of SLC34A1 mutations. Physiological Reports, 2018, 6, e13715.	0.7	32
26	Update on Hereditary Kidney Stone Disease and Introduction of a New Clinical Patient Registry in Germany. Frontiers in Pediatrics, 2018, 6, 47.	0.9	14
27	Biallelic Expression of Mucin-1 in Autosomal Dominant Tubulointerstitial Kidney Disease: Implications for Nongenetic Disease Recognition. Journal of the American Society of Nephrology: JASN, 2018, 29, 2298-2309.	3.0	25
28	Early Flow Disturbances of Tunnelled Haemodialysis Catheters and Topographic Landmarks in Chest X-Ray. Blood Purification, 2018, 46, 70-76.	0.9	0
29	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.	1.6	13
30	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. American Journal of Human Genetics, 2017, 100, 323-333.	2.6	29
31	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. CKJ: Clinical Kidney Journal, 2017, 10, 742-746.	1.4	5
32	Mutations in SLC26A1 Cause Nephrolithiasis. American Journal of Human Genetics, 2016, 98, 1228-1234.	2.6	41
33	FAT1 mutations cause a glomerulotubular nephropathy. Nature Communications, 2016, 7, 10822.	5.8	99
34	Diagnosing FSGS without kidney biopsy â€“ a novel INF2-mutation in a family with ESRD of unknown origin. BMC Medical Genetics, 2016, 17, 73.	2.1	16
35	Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. Clinical Journal of the American Society of Nephrology: CJASN, 2016, 11, 664-672.	2.2	105
36	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. Journal of Medical Genetics, 2016, 53, 208-214.	1.5	39

#	Article	IF	Citations
37	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.	2.6	74
38	Targeted Resequencing of 29 Candidate Genes and Mouse Expression Studies ImplicateZIC3andFOXF1in Human VATER/VACTERL Association. Human Mutation, 2015, 36, 1150-1154.	1.1	46
39	DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. American Journal of Human Genetics, 2015, 96, 81-92.	2.6	98
40	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	2.3	95
41	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665.	1.5	32
42	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.	3.0	499
43	Fourteen Monogenic Genes Account for 15% of Nephrolithiasis/Nephrocalcinosis. Journal of the American Society of Nephrology: JASN, 2015, 26, 543-551.	3.0	163
44	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.	2.2	74
45	Mutations in RSPH1 Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. American Journal of Respiratory and Critical Care Medicine, 2014, 189, 707-717.	2.5	191
46	Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86.	2.6	75
47	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. American Journal of Human Genetics, 2014, 94, 905-914.	2.6	90
48	Identification of 99 novel mutations in a worldwide cohort of 1,056 patients with a nephronophthisis-related ciliopathy. Human Genetics, 2013, 132, 865-884.	1.8	199
49	ANKS6 is a central component of a nephronophthisis module linking NEK8 to INVS and NPHP3. Nature Genetics, 2013, 45, 951-956.	9.4	183
50	ZMYND10 Is Mutated in Primary Ciliary Dyskinesia and Interacts with LRRC6. American Journal of Human Genetics, 2013, 93, 336-345.	2.6	183
51	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.	2.6	184
52	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. American Journal of Human Genetics, 2013, 93, 711-720.	2.6	135
53	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	2.6	196
54	Mutation of the Mg2+ Transporter SLC41A1 Results in a Nephronophthisis-Like Phenotype. Journal of the American Society of Nephrology: JASN, 2013, 24, 967-977.	3.0	63

#	Article	IF	Citations
55	THOC5: a novel gene involved in HDL-cholesterol metabolism. Journal of Lipid Research, 2013, 54, 3170-3176.	2.0	15
56	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.	1.5	109
57	Successful Simultaneous Pancreas Kidney Transplantation in Maturity-Onset Diabetes of the Young Type 5. Transplantation, 2011, 92, e45-e47.	0.5	3
58	Role of genetic variation in the human sodiumâ€“glucose cotransporter 2 gene (SGLT2) in glucose homeostasis. Pharmacogenomics, 2011, 12, 1119-1126.	0.6	37
59	Genetic and Evolutionary Analyses of the Human Bone Morphogenetic Protein Receptor 2 (BMPR2) in the Pathophysiology of Obesity. PLoS ONE, 2011, 6, e16155.	1.1	38
60	Isolated Renal Relapse of Sarcoidosis under Low-Dose Glucocorticoid Therapy. Journal of General Internal Medicine, 2008, 23, 879-882.	1.3	7
61	Novel somatic PBX1 mosaicism likely masking syndromic CAKUT in an adult with bilateral kidney hypoplasia. CKJ: Clinical Kidney Journal, 0, , .	1.4	2