Bodo B Beck

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7418061/publications.pdf

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56	2,278	257450	223800
papers	citations	24 h-index	46 g-index

57	57	57	2736
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	MAGED2 controls vasopressin-induced aquaporin-2 expression in collecting duct cells. Journal of Proteomics, 2022, 252, 104424.	2.4	1
2	Lifelong effect of therapy in young patients with the COL4A5 Alport missense variant p.(Gly624Asp): a prospective cohort study. Nephrology Dialysis Transplantation, 2022, 37, 2496-2504.	0.7	16
3	Unraveling Structural Rearrangements of the CFH Gene Cluster in Atypical Hemolytic Uremic Syndrome Patients Using Molecular Combing and Long-Fragment Targeted Sequencing. Journal of Molecular Diagnostics, 2022, 24, 619-631.	2.8	5
4	Diverse molecular causes of unsolved autosomal dominant tubulointerstitial kidney diseases. Kidney International, 2022, 102, 405-420.	5.2	10
5	Consensus draft of the native mouse podocyte-ome. American Journal of Physiology - Renal Physiology, 2022, 323, F182-F197.	2.7	6
6	Arterial Hypertension in a 10-Year-Old Girl. American Journal of Kidney Diseases, 2021, 77, A11-A13.	1.9	0
7	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
8	MO043HYPERURICEMIA IS RELATIVELY COMMON IN CHILDREN WITH HNF1B MUTATION, BUT ITS UTILITY AS A CLINICALLY USEFUL MARKER FOR PREDICTING THE MUTATION IS LIMITED. Nephrology Dialysis Transplantation, 2021, 36, .	0.7	2
9	Expanding the Spectrum of FAT1 Nephropathies by Novel Mutations That Affect Hippo Signaling. Kidney International Reports, 2021, 6, 1368-1378.	0.8	7
10	A new era of treatment for primary hyperoxaluria type 1. Nature Reviews Nephrology, 2021, 17, 573-574.	9.6	5
11	NPHP1 gene-associated nephronophthisis is associated with an occult retinopathy. Kidney International, 2021, 100, 1092-1100.	5.2	6
12	Hyperuricemia Is an Early and Relatively Common Feature in Children with HNF1B Nephropathy but Its Utility as a Predictor of the Disease Is Limited. Journal of Clinical Medicine, 2021, 10, 3265.	2.4	7
13	A report from the European Hyperoxaluria Consortium (OxalEurope) Registry on a large cohort of patients with primary hyperoxaluria type 3. Kidney International, 2021, 100, 621-635.	5.2	26
14	Refining genotypeâ€“phenotype correlations in 304 patients with autosomal recessive polycystic kidney disease and PKHD1 gene variants. Kidney International, 2021, 100, 650-659.	5.2	38
15	Four Cases of Maturity Onset Diabetes of the Young (MODY) Type 5 Associated with Mutations in the Hepatocyte Nuclear Factor 1 Beta (HNF1B) Gene Presenting in a 13-Year-Old Boy and in Adult Men Aged 33, 34, and 35 Years in Poland. American Journal of Case Reports, 2021, 22, e928994.	0.8	6
16	mTOR-Activating Mutations in RRAGD Are Causative for Kidney Tubulopathy and Cardiomyopathy. Journal of the American Society of Nephrology: JASN, 2021, 32, 2885-2899.	6.1	24
17	Clinical profile of a Polish cohort of children and young adults with cystinuria. Renal Failure, 2021, 43, 62-70.	2.1	2
18	An international cohort study of autosomal dominant tubulointerstitial kidney disease due to mutations identifies distinct clinical subtypes. Kidney International, 2020, 98, 1589-1604.	5.2	27

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19	Hypomagnesemia is underestimated in children with HNF1B mutations. Pediatric Nephrology, 2020, 35, 1877-1886.	1.7	11
20	A molecular mechanism explaining albuminuria in kidney disease. Nature Metabolism, 2020, 2, 461-474.	11.9	99
21	Case Report: Exome Sequencing Reveals LRBA Deficiency in a Patient With End-Stage Renal Disease. Frontiers in Pediatrics, 2020, 8, 42.	1.9	2
22	A step towards precision medicine in management of severe transient polyhydramnios: MAGED2 variant. Journal of Obstetrics and Gynaecology, 2019, 39, 395-397.	0.9	6
23	Generation of an induced pluripotent stem cell line (CIMAi001-A) from a compound heterozygous Primary Hyperoxaluria Type I (PH1) patient carrying p.G170R and p.R122* mutations in the AGXT gene.. Stem Cell Research, 2019, 41, 101626.	0.7	3
24	Subclinical myocardial disease in patients with primary hyperoxaluria and preserved left ventricular ejection fraction: a two-dimensional speckle-tracking imaging study. Pediatric Nephrology, 2019, 34, 2591-2600.	1.7	11
25	Patients with primary hyperoxaluria type 2 have significant morbidity and require careful follow-up. Kidney International, 2019, 96, 1389-1399.	5.2	61
26	Autosomal Tubulointerstitial Kidney Diseaseâ€"MUC1 Type: Differential Proteomics Suggests that Mutated MUC1 (insC) Affects Vesicular Transport in Renal Epithelial Cells. Proteomics, 2018, 18, e1700456.	2.2	13
27	Single-nephron proteomes connect morphology and function in proteinuric kidney disease. Kidney International, 2018, 93, 1308-1319.	5.2	49
28	Single molecule real time sequencing in ADTKD-MUC1 allows complete assembly of the VNTR and exact positioning of causative mutations. Scientific Reports, 2018, 8, 4170.	3.3	40
29	EinfÃ¼hrung zum Thema: Erbliche Nierenerkrankungen. Medizinische Genetik, 2018, 30, 389-390.	0.2	0
30	Steroid-resistentes nephrotisches Syndrom. Medizinische Genetik, 2018, 30, 410-421.	0.2	3
31	Genetische Nierensteinerkrankungen. Medizinische Genetik, 2018, 30, 438-447.	0.2	0
32	Germline De Novo Mutations in ATP1A1 Cause Renal Hypomagnesemia, Refractory Seizures, and Intellectual Disability. American Journal of Human Genetics, 2018, 103, 808-816.	6.2	74
33	A de novo <i>KCNA1</i> Mutation in a Patient with Tetany and Hypomagnesemia. Nephron, 2018, 139, 359-366.	1.8	22
34	Noninvasive Immunohistochemical Diagnosis and Novel MUC1 Mutations Causing Autosomal Dominant Tubulointerstitial Kidney Disease. Journal of the American Society of Nephrology: JASN, 2018, 29, 2418-2431.	6.1	38
35	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.	6.1	25
36	Renal thrombotic microangiopathy in patients with cblC defect: review of an under-recognized entity. Pediatric Nephrology, 2017, 32, 733-741.	1.7	76

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37	Systematic assessment of urinary hydroxy-oxo-glutarate for diagnosis and follow-up of primary hyperoxaluria type III. Pediatric Nephrology, 2017, 32, 2263-2271.	1.7	22
38	Characterization of a splice-site mutation in the tumor suppressor gene FLCN associated with renal cancer. BMC Medical Genetics, 2017, 18, 53.	2.1	13
39	Mucin-1 Increases Renal TRPV5 Activity In Vitro, and Urinary Level Associates with Calcium Nephrolithiasis in Patients. Journal of the American Society of Nephrology: JASN, 2016, 27, 3447-3458.	6.1	38
40	Polyhydramnios, Transient Antenatal Bartterâ€™s Syndrome, andMAGED2Mutations. New England Journal of Medicine, 2016, 374, 1853-1863.	27.0	148
41	Familial juvenile hyperuricemic nephropathy as rare cause of dialysis-dependent chronic kidney diseaseâ€"a series of cases in two families. Renal Failure, 2016, 38, 1759-1762.	2.1	9
42	Rapid Response to Cyclosporin A and Favorable Renal Outcome in Nongenetic Versus Genetic Steroidâ€"Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2016, 11, 245-253.	4.5	103
43	Three-layered proteomic characterization of a novelACTN4mutation unravels its pathogenic potential in FSGS. Human Molecular Genetics, 2016, 25, 1152-1164.	2.9	36
44	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
45	Primary hyperoxaluria â€" An update. Journal of Pediatric Biochemistry, 2015, 04, 101-110.	0.2	0
46	Inhomogeneous Longitudinal Cardiac Rotation and Impaired Left Ventricular Longitudinal Strain in Children and Young Adults with Endâ€Stage Renal Failure Undergoing Hemodialysis. Echocardiography, 2015, 32, 1250-1260.	0.9	11
47	Renal fibrosis is the common feature of autosomal dominant tubulointerstitial kidney diseases caused by mutations in mucin 1 or uromodulin. Kidney International, 2014, 86, 589-599.	5.2	86
48	Data from a large European study indicate that the outcome of primary hyperoxaluria type 1 correlates with the AGXT mutation type. Kidney International, 2014, 86, 1197-1204.	5.2	103
49	Vitamin B6 in Primary Hyperoxaluria I. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 468-477.	4.5	110
50	Hyperoxaluria and systemic oxalosis: an update on current therapy and future directions. Expert Opinion on Investigational Drugs, 2013, 22, 117-129.	4.1	61
51	Novel findings in patients with primary hyperoxaluria type III and implications for advanced molecular testing strategies. European Journal of Human Genetics, 2013, 21, 162-172.	2.8	71
52	ARHGDIA mutations cause nephrotic syndrome via defective RHO GTPase signaling. Journal of Clinical Investigation, 2013, 123, 3243-3253.	8.2	196
53	Liver cell transplantation in severe infantile oxalosis--a potential bridging procedure to orthotopic liver transplantation?. Nephrology Dialysis Transplantation, 2012, 27, 2984-2989.	0.7	43
54	Autosomal Dominant Mutation in the Signal Peptide of Renin in a Kindred With Anemia, Hyperuricemia, and CKD. American Journal of Kidney Diseases, 2011, 58, 821-825.	1.9	26

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55	C3 deposition glomerulopathy due to a functional Factor H defect. Kidney International, 2009, 75, 1230-1234.	5.2	79
56	The primary hyperoxalurias. Kidney International, 2009, 75, 1264-1271.	5.2	314