

Julia Hoefele

List of Publications by Year in descending order

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Version: 2024-02-01

78
papers

3,442
citations

249298

26
h-index

169272

56
g-index

84
all docs

84
docs citations

84
times ranked

4262
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in INVS encoding inversin cause nephronophthisis type 2, linking renal cystic disease to the function of primary cilia and left-right axis determination. <i>Nature Genetics</i> , 2003, 34, 413-420.	9.4	582
2	Mutations in a novel gene, NPHP3, cause adolescent nephronophthisis, tapeto-retinal degeneration and hepatic fibrosis. <i>Nature Genetics</i> , 2003, 34, 455-459.	9.4	345
3	Mutations in Multiple PKD Genes May Explain Early and Severe Polycystic Kidney Disease. <i>Journal of the American Society of Nephrology: JASN</i> , 2011, 22, 2047-2056.	3.0	211
4	ARHGDI1 mutations cause nephrotic syndrome via defective RHO GTPase signaling. <i>Journal of Clinical Investigation</i> , 2013, 123, 3243-3253.	3.9	196
5	A Gene Mutated in Nephronophthisis and Retinitis Pigmentosa Encodes a Novel Protein, Nephroretinin, Conserved in Evolution. <i>American Journal of Human Genetics</i> , 2002, 71, 1161-1167.	2.6	193
6	KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2015, 125, 2375-2384.	3.9	159
7	Evidence of Oligogenic Inheritance in Nephronophthisis. <i>Journal of the American Society of Nephrology: JASN</i> , 2007, 18, 2789-2795.	3.0	141
8	Rapid Response to Cyclosporin A and Favorable Renal Outcome in Nongenetic Versus Genetic Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2016, 11, 245-253.	2.2	103
9	Genotype-phenotype correlation in 440 patients with NPHP-related ciliopathies. <i>Kidney International</i> , 2011, 80, 1239-1245.	2.6	99
10	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. <i>Pediatric Nephrology</i> , 2019, 34, 1175-1189.	0.9	97
11	Identification of two novel CAKUT-causing genes by massively parallel exon resequencing of candidate genes in patients with unilateral renal agenesis. <i>Kidney International</i> , 2012, 81, 196-200.	2.6	75
12	Phenotypic features of carbohydrate sulfotransferase 3 (CHST3) deficiency in 24 patients: Congenital dislocations and vertebral changes as principal diagnostic features. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2543-2549.	0.7	67
13	<i>De novo</i> variants in neurodevelopmental disorders—experiences from a tertiary care center. <i>Clinical Genetics</i> , 2021, 100, 14-28.	1.0	64
14	Consensus statement on standards and guidelines for the molecular diagnostics of Alport syndrome: refining the ACMG criteria. <i>European Journal of Human Genetics</i> , 2021, 29, 1186-1197.	1.4	61
15	Mutational analysis of the NPHP4 gene in 250 patients with nephronophthisis. <i>Human Mutation</i> , 2005, 25, 411-411.	1.1	60
16	Mapping of Gene Loci for Nephronophthisis Type 4 and Senior-Løken Syndrome, to Chromosome 1p36. <i>American Journal of Human Genetics</i> , 2002, 70, 1240-1246.	2.6	56
17	Guidelines for Genetic Testing and Management of Alport Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2022, 17, 143-154.	2.2	49
18	Metabolic control analysis of the Warburg-effect in proliferating vascular smooth muscle cells. <i>Journal of Biomedical Science</i> , 2005, 12, 827-834.	2.6	48

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19	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
20	Histone H3.3 beyond cancer: Germline mutations in <i>Histone 3 Family 3A and 3B</i> cause a previously unidentified neurodegenerative disorder in 46 patients. <i>Science Advances</i> , 2020, 6, .	4.7	43
21	Characterization of SETD1A haploinsufficiency in humans and <i>Drosophila</i> defines a novel neurodevelopmental syndrome. <i>Molecular Psychiatry</i> , 2021, 26, 2013-2024.	4.1	43
22	Multisystem inflammation and susceptibility to viral infections in human ZNFX1 deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 381-393.	1.5	40
23	Novel PKD1 and PKD2 mutations in autosomal dominant polycystic kidney disease (ADPKD). <i>Nephrology Dialysis Transplantation</i> , 2011, 26, 2181-2188.	0.4	36
24	Next generation sequencing as a useful tool in the diagnostics of mosaicism in Alport syndrome. <i>Gene</i> , 2013, 526, 474-477.	1.0	33
25	Exome Sequencing and Identification of Phenocopies in Patients With Clinically Presumed Hereditary Nephropathies. <i>American Journal of Kidney Diseases</i> , 2020, 76, 460-470.	2.1	33
26	Identification of 47 novel mutations in patients with Alport syndrome and thin basement membrane nephropathy. <i>Pediatric Nephrology</i> , 2016, 31, 941-955.	0.9	32
27	Pseudodominant inheritance of nephronophthisis caused by a homozygous NPHP1 deletion. <i>Pediatric Nephrology</i> , 2011, 26, 967-971.	0.9	26
28	Expanding the mutation spectrum for Fraser syndrome: Identification of a novel heterozygous deletion in FRAS1. <i>Gene</i> , 2013, 520, 194-197.	1.0	24
29	Missense mutations in EYA1 and TCF2 are a rare cause of urinary tract malformations. <i>Nephrology Dialysis Transplantation</i> , 2007, 23, 777-779.	0.4	23
30	Multiple urinary tract malformations with likely recessive inheritance in a large Somalian kindred. <i>Nephrology Dialysis Transplantation</i> , 2004, 19, 3172-3175.	0.4	22
31	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. <i>Genetics in Medicine</i> , 2021, 23, 1715-1725.	1.1	22
32	Dealing with the incidental finding of secondary variants by the example of SRNS patients undergoing targeted next-generation sequencing. <i>Pediatric Nephrology</i> , 2016, 31, 73-81.	0.9	19
33	A case report and review of the literature indicate that HMGA2 should be added as a disease gene for Silver-Russell syndrome. <i>Gene</i> , 2018, 663, 110-114.	1.0	18
34	Blood DNA methylation provides an accurate biomarker of <i>KMT2B</i> -related dystonia and predicts onset. <i>Brain</i> , 2022, 145, 644-654.	3.7	18
35	Mapping of a new locus for congenital anomalies of the kidney and urinary tract on chromosome 8q24. <i>Nephrology Dialysis Transplantation</i> , 2010, 25, 1496-1501.	0.4	17
36	Oral Coenzyme Q10 supplementation leads to better preservation of kidney function in steroid-resistant nephrotic syndrome due to primary Coenzyme Q10 deficiency. <i>Kidney International</i> , 2022, 102, 604-612.	2.6	17

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37	Heterozygous COL4A3 Variants in Histologically Diagnosed Focal Segmental Glomerulosclerosis. <i>Frontiers in Pediatrics</i> , 2018, 6, 171.	0.9	16
38	Lifelong effect of therapy in young patients with the <i>COL4A5</i> Alport missense variant p.(Gly624Asp): a prospective cohort study. <i>Nephrology Dialysis Transplantation</i> , 2022, 37, 2496-2504.	0.4	16
39	Novel heterozygous COL4A3 mutation in a family with late-onset ESRD. <i>Pediatric Nephrology</i> , 2010, 25, 1539-1542.	0.9	15
40	Identification of a Novel Heterozygous De Novo 7-bp Frameshift Deletion in PBX1 by Whole-Exome Sequencing Causing a Multi-Organ Syndrome Including Bilateral Dysplastic Kidneys and Hypoplastic Clavicles. <i>Frontiers in Pediatrics</i> , 2017, 5, 251.	0.9	14
41	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. <i>American Journal of Human Genetics</i> , 2021, 108, 357-367.	2.6	14
42	Mutational analysis in 119 families with nephronophthisis. <i>Pediatric Nephrology</i> , 2007, 22, 366-370.	0.9	13
43	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. <i>European Journal of Human Genetics</i> , 2021, 29, 411-421.	1.4	13
44	DAAM2 Variants Cause Nephrotic Syndrome via Actin Dysregulation. <i>American Journal of Human Genetics</i> , 2020, 107, 1113-1128.	2.6	12
45	The 2019 and 2021 International Workshops on Alport Syndrome. <i>European Journal of Human Genetics</i> , 2022, 30, 507-516.	1.4	12
46	Variation of the clinical spectrum and genotype-phenotype associations in Coenzyme Q10 deficiency associated glomerulopathy. <i>Kidney International</i> , 2022, 102, 592-603.	2.6	12
47	A De Novo Missense Variant in POU3F2 Identified in a Child with Global Developmental Delay. <i>Neuropediatrics</i> , 2018, 49, 401-404.	0.3	11
48	The Hypomorphic Variant p.(Gly624Asp) in COL4A5 as a Possible Cause for an Unexpected Severe Phenotype in a Family With X-Linked Alport Syndrome. <i>Frontiers in Pediatrics</i> , 2019, 7, 485.	0.9	11
49	Identification of disease-causing variants by comprehensive genetic testing with exome sequencing in adults with suspicion of hereditary FSGS. <i>European Journal of Human Genetics</i> , 2021, 29, 262-270.	1.4	11
50	Clinical and histological presentation of 3 siblings with mutations in the NPHP4 gene. <i>American Journal of Kidney Diseases</i> , 2004, 43, 358-364.	2.1	10
51	Foreign body in the bladder mimicking nephritis. <i>Pediatric Nephrology</i> , 2007, 22, 467-470.	0.9	10
52	Truncating <i>WT1</i> Mutation in an XX Female with Adult-Onset Focal Segmental Glomerulosclerosis and Streak Ovaries: A Case Report. <i>Nephron</i> , 2017, 135, 72-76.	0.9	10
53	A novel interstitial deletion of 10q24.2q24.32 in a patient with renal coloboma syndrome. <i>European Journal of Medical Genetics</i> , 2012, 55, 211-215.	0.7	9
54	Identification of a de novo microdeletion 1q44 in a patient with hypogenesis of the corpus callosum, seizures and microcephaly – A case report. <i>Gene</i> , 2017, 616, 41-44.	1.0	9

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55	Mutations in INF2 may be associated with renal histology other than focal segmental glomerulosclerosis. <i>Pediatric Nephrology</i> , 2018, 33, 433-437.	0.9	9
56	MAP2 â€“ A Candidate Gene for Epilepsy, Developmental Delay and Behavioral Abnormalities in a Patient With Microdeletion 2q34. <i>Frontiers in Genetics</i> , 2018, 9, 99.	1.1	9
57	Identification of co-occurrence in a patient with Dent's disease and ADA2-deficiency by exome sequencing. <i>Gene</i> , 2018, 649, 23-26.	1.0	8
58	COL4A5-associated X-linked Alport syndrome in a female patient with early inner ear deafness due to a mutation in MYH9. <i>Nephrology Dialysis Transplantation</i> , 2012, 27, 4236-4240.	0.4	7
59	Precise variant interpretation, phenotype ascertainment, and genotypeâ€“phenotype correlation of children in the <scp>EARLY PROâ€“TECT</scp> Alport trial. <i>Clinical Genetics</i> , 2021, 99, 143-156.	1.0	7
60	Clonal hematopoiesis as a pitfall in germline variant interpretation in the context of Mendelian disorders. <i>Human Molecular Genetics</i> , 2022, 31, 2386-2395.	1.4	7
61	Haemophilus paraphrophilus, a rare cause of intracerebral abscess in children. <i>European Journal of Pediatrics</i> , 2008, 167, 629-632.	1.3	6
62	Congenital lymphedema as a rare and first symptom of tuberous sclerosis complex. <i>Gene</i> , 2020, 753, 144815.	1.0	6
63	A novel pathogenic variant in MYO18B associating early-onset muscular hypotonia, and characteristic dysmorphic features, delineation of the phenotypic spectrum of MYO18B-related conditions. <i>Gene</i> , 2020, 742, 144542.	1.0	6
64	Reâ€“sequencing of candidate genes <scp>FOXF1</scp>, <scp>HSPA6</scp>, <scp>HAAO</scp>, and <scp>KYNU</scp> in 522 individuals with <scp>VATER</scp>/<scp>VACTERL</scp>, <scp>VACTER</scp>/<scp>VACTERL</scp>â€“like association, and isolated anorectal malformation. <i>Birth Defects Research</i> , 2022, 114, 478-486.	0.8	6
65	Cyclosporine A responsive congenital nephrotic syndrome with single heterozygous variants in NPHS1, NPHS2, and PLCE1. <i>Pediatric Nephrology</i> , 2018, 33, 1269-1272.	0.9	5
66	Refining Kidney Survival in 383 Genetically Characterized Patients With Nephronophthisis. <i>Kidney International Reports</i> , 2022, 7, 2016-2028.	0.4	5
67	Disorders of sex development and Diamond-Blackfan anemia: is there an association?. <i>Pediatric Nephrology</i> , 2010, 25, 1255-1261.	0.9	4
68	Mosaic trisomy 12 diagnosed in a female patient: clinical features, genetic analysis, and review of the literature. <i>World Journal of Pediatrics</i> , 2021, 17, 438-448.	0.8	4
69	Steroid-resistentes nephrotisches Syndrom. <i>Medizinische Genetik</i> , 2018, 30, 410-421.	0.1	3
70	BK virus induced nephritis in a boy with acute myeloid leukaemia undergoing bone marrow transplantation. <i>CKJ: Clinical Kidney Journal</i> , 2008, 1, 336-339.	1.4	2
71	Reduced Methotrexate Clearance and Renal Impairment in a Boy With Osteosarcoma and Earlier Undetected Autosomal Dominant Polycystic Kidney Disease (ADPKD). <i>Journal of Pediatric Hematology/Oncology</i> , 2010, 32, e314-e316.	0.3	2
72	Different factor H-related protein patterns in siblings with typical hemolytic uremic syndrome. <i>Pediatric Nephrology</i> , 2011, 26, 1345-1347.	0.9	2

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73	Rare co-occurrence of osteogenesis imperfecta type I and autosomal dominant polycystic kidney disease. <i>World Journal of Pediatrics</i> , 2016, 12, 501-503.	0.8	2
74	Correlation of PET-MRI, pathology, LOH and surgical success in a case of CHI with atypical large pancreatic focus. <i>Journal of the Endocrine Society</i> , 2022, 6, bvac056.	0.1	2
75	Renal and Skeletal Anomalies in a Cohort of Individuals With Clinically Presumed Hereditary Nephropathy Analyzed by Molecular Genetic Testing. <i>Frontiers in Genetics</i> , 2021, 12, 642849.	1.1	1
76	Exome sequencing in individuals with cardiovascular laterality defects identifies potential candidate genes. <i>European Journal of Human Genetics</i> , 2022, , .	1.4	1
77	No Impact of the Analytical Method Used for Determining Cystatin C on Estimating Glomerular Filtration Rate in Children. <i>Frontiers in Pediatrics</i> , 2017, 5, 66.	0.9	0
78	Genetische Ursachen und Therapie beim Alport-Syndrom. <i>Medizinische Genetik</i> , 2019, 30, 429-437.	0.1	0