Julia Hoefele

List of Publications by Year
in descending order

78	3,442	249298 26	169272 56
papers	citations	h-index	g-index

84	84	84	4262
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Blood DNA methylation provides an accurate biomarker of KMT2B-related dystonia and predicts onset. Brain, 2022, 145, 644-654.	3.7	18
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.	0.4	16
3	Clonal hematopoiesis as a pitfall in germline variant interpretation in the context of Mendelian disorders. Human Molecular Genetics, 2022, 31, 2386-2395.	1.4	7
4	The 2019 and 2021 International Workshops on Alport Syndrome. European Journal of Human Genetics, 2022, 30, 507-516.	1.4	12
5	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. Birth Defects Research, 2022, 114, 478-486.	0.8	6
6	Correlation of PET-MRI, pathology, LOH and surgical success in a case of CHI with atypical large pancreatic focus. Journal of the Endocrine Society, 2022, 6, bvac056.	0.1	2
7	Guidelines for Genetic Testing and Management of Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2022, 17, 143-154.	2.2	49
8	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
9	Variation of the clinical spectrum and genotype-phenotype associations in Coenzyme Q10 deficiency associated glomerulopathy. Kidney International, 2022, 102, 592-603.	2.6	12
10	Exome sequencing in individuals with cardiovascular laterality defects identifies potential candidate genes. European Journal of Human Genetics, 2022, , .	1.4	1
11	Oral Coenzyme Q10 supplementation leads to better preservation of kidney function in steroid-resistant nephrotic syndrome due to primary Coenzyme Q10 deficiency. Kidney International, 2022, 102, 604-612.	2.6	17
12	Refining Kidney Survival in 383 Genetically Characterized Patients With Nephronophthisis. Kidney International Reports, 2022, 7, 2016-2028.	0.4	5
13	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	4.1	43
14	Precise variant interpretation, phenotype ascertainment, and genotypeâ€“phenotype correlation of children in the <scp>EARLY PROâ€TECT</scp> Alport trial. Clinical Genetics, 2021, 99, 143-156.	1.0	7
15	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. European Journal of Human Genetics, 2021, 29, 411-421.	1.4	13
16	Identification of disease-causing variants by comprehensive genetic testing with exome sequencing in adults with suspicion of hereditary FSGS. European Journal of Human Genetics, 2021, 29, 262-270.	1.4	11
17	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.	2.6	14
18	De novo variants in neurodevelopmental disordersâ€”experiences from a tertiary care center. Clinical Genetics, 2021, 100, 14-28.	1.0	64

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19	Consensus statement on standards and guidelines for the molecular diagnostics of Alport syndrome: refining the ACMG criteria. European Journal of Human Genetics, 2021, 29, 1186-1197.	1.4	61
20	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	1.1	22
21	Renal and Skeletal Anomalies in a Cohort of Individuals With Clinically Presumed Hereditary Nephropathy Analyzed by Molecular Genetic Testing. Frontiers in Genetics, 2021, 12, 642849.	1.1	1
22	Mosaic trisomy 12 diagnosed in a female patient: clinical features, genetic analysis, and review of the literature. World Journal of Pediatrics, 2021, 17, 438-448.	0.8	4
23	Multisystem inflammation and susceptibility to viral infections in human ZNFX1 deficiency. Journal of Allergy and Clinical Immunology, 2021, 148, 381-393.	1.5	40
24	DAAM2 Variants Cause Nephrotic Syndrome via Actin Dysregulation. American Journal of Human Genetics, 2020, 107, 1113-1128.	2.6	12
25	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
26	Exome Sequencing and Identification of Phenocopies in Patients With Clinically Presumed Hereditary Nephropathies. American Journal of Kidney Diseases, 2020, 76, 460-470.	2.1	33
27	Congenital lymphedema as a rare and first symptom of tuberous sclerosis complex. Gene, 2020, 753, 144815.	1.0	6
28	A novel pathogenic variant in MYO18B associating early-onset muscular hypotonia, and characteristic dysmorphic features, delineation of the phenotypic spectrum of MYO18B-related conditions. Gene, 2020, 742, 144542.	1.0	6
29	Genetische Ursachen und Therapie beim Alport-Syndrom. Medizinische Genetik, 2019, 30, 429-437.	0.1	0
30	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. Pediatric Nephrology, 2019, 34, 1175-1189.	0.9	97
31	The Hypomorphic Variant p.(Gly624Asp) in COL4A5 as a Possible Cause for an Unexpected Severe Phenotype in a Family With X-Linked Alport Syndrome. Frontiers in Pediatrics, 2019, 7, 485.	0.9	11
32	Cyclosporine A responsive congenital nephrotic syndrome with single heterozygous variants in NPHS1, NPHS2, and PLCE1. Pediatric Nephrology, 2018, 33, 1269-1272.	0.9	5
33	A case report and review of the literature indicate that HMGA2 should be added as a disease gene for Silver-Russell syndrome. Gene, 2018, 663, 110-114.	1.0	18
34	Identification of co-occurrence in a patient with Dent's disease and ADA2-deficiency by exome sequencing. Gene, 2018, 649, 23-26.	1.0	8
35	Mutations in INF2 may be associated with renal histology other than focal segmental glomerulosclerosis. Pediatric Nephrology, 2018, 33, 433-437.	0.9	9
36	Steroid-resistentes nephrotisches Syndrom. Medizinische Genetik, 2018, 30, 410-421.	0.1	3

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37	A De Novo Missense Variant in POU3F2 Identified in a Child with Global Developmental Delay. Neuropediatrics, 2018, 49, 401-404.	0.3	11
38	Heterozygous COL4A3 Variants in Histologically Diagnosed Focal Segmental Glomerulosclerosis. Frontiers in Pediatrics, 2018, 6, 171.	0.9	16
39	MAP2 â€“ A Candidate Gene for Epilepsy, Developmental Delay and Behavioral Abnormalities in a Patient With Microdeletion 2q34. Frontiers in Genetics, 2018, 9, 99.	1.1	9
40	Truncating <i>Wilms Tumor Suppressor Gene 1</i> Mutation in an XX Female with Adult-Onset Focal Segmental Glomerulosclerosis and Streak Ovaries: A Case Report. Nephron, 2017, 135, 72-76.	0.9	10
41	Identification of a de novo microdeletion 1q44 in a patient with hypogenesis of the corpus callosum, seizures and microcephaly â€“ A case report. Gene, 2017, 616, 41-44.	1.0	9
42	No Impact of the Analytical Method Used for Determining Cystatin C on Estimating Glomerular Filtration Rate in Children. Frontiers in Pediatrics, 2017, 5, 66.	0.9	0
43	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. Frontiers in Pediatrics, 2017, 5, 251.	0.9	14
44	Rare co-occurrence of osteogenesis imperfecta type I and autosomal dominant polycystic kidney disease. World Journal of Pediatrics, 2016, 12, 501-503.	0.8	2
45	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.	2.2	103
46	Identification of 47 novel mutations in patients with Alport syndrome and thin basement membrane nephropathy. Pediatric Nephrology, 2016, 31, 941-955.	0.9	32
47	Dealing with the incidental finding of secondary variants by the example of SRNS patients undergoing targeted next-generation sequencing. Pediatric Nephrology, 2016, 31, 73-81.	0.9	19
48	KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. Journal of Clinical Investigation, 2015, 125, 2375-2384.	3.9	159
49	Next generation sequencing as a useful tool in the diagnostics of mosaicism in Alport syndrome. Gene, 2013, 526, 474-477.	1.0	33
50	Expanding the mutation spectrum for Fraser syndrome: Identification of a novel heterozygous deletion in FRAS1. Gene, 2013, 520, 194-197.	1.0	24
51	ARHGDIA mutations cause nephrotic syndrome via defective RHO GTPase signaling. Journal of Clinical Investigation, 2013, 123, 3243-3253.	3.9	196
52	Identification of two novel CAKUT-causing genes by massively parallel exon resequencing of candidate genes in patients with unilateral renal agenesis. Kidney International, 2012, 81, 196-200.	2.6	75
53	COL4A5-associated X-linked Alport syndrome in a female patient with early inner ear deafness due to a mutation in MYH9. Nephrology Dialysis Transplantation, 2012, 27, 4236-4240.	0.4	7
54	A novel interstitial deletion of 10q24.2q24.32 in a patient with renal coloboma syndrome. European Journal of Medical Genetics, 2012, 55, 211-215.	0.7	9

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55	Genotypeâ€“phenotype correlation in 440 patients with NPHP-related ciliopathies. Kidney International, 2011, 80, 1239-1245.	2.6	99
56	Pseudodominant inheritance of nephronophthisis caused by a homozygous NPHP1 deletion. Pediatric Nephrology, 2011, 26, 967-971.	0.9	26
57	Different factor H-related protein patterns in siblings with typical hemolytic uremic syndrome. Pediatric Nephrology, 2011, 26, 1345-1347.	0.9	2
58	Novel PKD1 and PKD2 mutations in autosomal dominant polycystic kidney disease (ADPKD). Nephrology Dialysis Transplantation, 2011, 26, 2181-2188.	0.4	36
59	Mutations in Multiple PKD Genes May Explain Early and Severe Polycystic Kidney Disease. Journal of the American Society of Nephrology: JASN, 2011, 22, 2047-2056.	3.0	211
60	Reduced Methotrexate Clearance and Renal Impairment in a Boy With Osteosarcoma and Earlier Undetected Autosomal Dominant Polycystic Kidney Disease (ADPKD). Journal of Pediatric Hematology/Oncology, 2010, 32, e314-e316.	0.3	2
61	Novel heterozygous COL4A3 mutation in a family with late-onset ESRD. Pediatric Nephrology, 2010, 25, 1539-1542.	0.9	15
62	Disorders of sex development and Diamond-Blackfan anemia: is there an association?. Pediatric Nephrology, 2010, 25, 1255-1261.	0.9	4
63	Phenotypic features of carbohydrate sulfotransferase 3 (CHST3) deficiency in 24 patients: Congenital dislocations and vertebral changes as principal diagnostic features. American Journal of Medical Genetics, Part A, 2010, 152A, 2543-2549.	0.7	67
64	Mapping of a new locus for congenital anomalies of the kidney and urinary tract on chromosome 8q24. Nephrology Dialysis Transplantation, 2010, 25, 1496-1501.	0.4	17
65	Haemophilus paraphrophilus, a rare cause of intracerebral abscess in children. European Journal of Pediatrics, 2008, 167, 629-632.	1.3	6
66	BK virus induced nephritis in a boy with acute myeloid leukaemia undergoing bone marrow transplantation. CKJ: Clinical Kidney Journal, 2008, 1, 336-339.	1.4	2
67	Evidence of Oligogenic Inheritance in Nephronophthisis. Journal of the American Society of Nephrology: JASN, 2007, 18, 2789-2795.	3.0	141
68	Missense mutations in EYA1 and TCF2 are a rare cause of urinary tract malformations. Nephrology Dialysis Transplantation, 2007, 23, 777-779.	0.4	23
69	Mutational analysis in 119 families with nephronophthisis. Pediatric Nephrology, 2007, 22, 366-370.	0.9	13
70	Foreign body in the bladder mimicking nephritis. Pediatric Nephrology, 2007, 22, 467-470.	0.9	10
71	Mutational analysis of theNPHP4 gene in 250 patients with nephronophthisis. Human Mutation, 2005, 25, 411-411.	1.1	60
72	Metabolic control analysis of the Warburg-effect in proliferating vascular smooth muscle cells. Journal of Biomedical Science, 2005, 12, 827-834.	2.6	48

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73	Multiple urinary tract malformations with likely recessive inheritance in a large Somalian kindred. Nephrology Dialysis Transplantation, 2004, 19, 3172-3175.	0.4	22
74	Clinical and histological presentation of 3 siblings with mutations in the NPHP4 gene. American Journal of Kidney Diseases, 2004, 43, 358-364.	2.1	10
75	Mutations in a novel gene, NPHP3, cause adolescent nephronophthisis, tapeto-retinal degeneration and hepatic fibrosis. Nature Genetics, 2003, 34, 455-459.	9.4	345
76	Mutations in INVS encoding inversin cause nephronophthisis type 2, linking renal cystic disease to the function of primary cilia and left-right axis determination. Nature Genetics, 2003, 34, 413-420.	9.4	582
77	Mapping of Gene Loci for Nephronophthisis Type 4 and Senior-LÃ¸ken Syndrome, to Chromosome 1p36. American Journal of Human Genetics, 2002, 70, 1240-1246.	2.6	56
78	A Gene Mutated in Nephronophthisis and Retinitis Pigmentosa Encodes a Novel Protein, Nephroretinin, Conserved in Evolution. American Journal of Human Genetics, 2002, 71, 1161-1167.	2.6	193