# Julia Hoefele

## List of Publications by Year in descending order



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|---|---|---|---|
| 78 papers | 3,442 citations | 218677 26 h-index | 149698 56 g-index |
| 84 all docs | 84 docs citations | 84 times ranked | 4012 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. Nature Genetics, 2003, 34, 413-420. | 21.4 | 582 |
| 2 | Mutations in a novel gene, NPHP3, cause adolescent nephronophthisis, tapeto-retinal degeneration and hepatic fibrosis. Nature Genetics, 2003, 34, 455-459. | 21.4 | 345 |
| 3 | 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. | 6.1 | 211 |
| 4 | ARHGDIA mutations cause nephrotic syndrome via defective RHO GTPase signaling. Journal of Clinical Investigation, 2013, 123, 3243-3253. | 8.2 | 196 |
| 5 | 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. | 6.2 | 193 |
| 6 | KANK deficiency leads to podocyte dysfunction and nephrotic syndrome. Journal of Clinical Investigation, 2015, 125, 2375-2384. | 8.2 | 159 |
| 7 | Evidence of Oligogenic Inheritance in Nephronophthisis. Journal of the American Society of Nephrology: JASN, 2007, 18, 2789-2795. | 6.1 | 141 |
| 8 | 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 |
| 9 | Genotypeâ€“phenotype correlation in 440 patients with NPHP-related ciliopathies. Kidney International, 2011, 80, 1239-1245. | 5.2 | 99 |
| 10 | Expert consensus guidelines for the genetic diagnosis of Alport syndrome. Pediatric Nephrology, 2019, 34, 1175-1189. | 1.7 | 97 |
| 11 | 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. | 5.2 | 75 |
| 12 | 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. | 1.2 | 67 |
| 13 | De novo variants in neurodevelopmental disordersâ€”experiences from a tertiary care center. Clinical Genetics, 2021, 100, 14-28. | 2.0 | 64 |
| 14 | 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. | 2.8 | 61 |
| 15 | Mutational analysis of theNPHP4 gene in 250 patients with nephronophthisis. Human Mutation, 2005, 25, 411-411. | 2.5 | 60 |
| 16 | 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. | 6.2 | 56 |
| 17 | Guidelines for Genetic Testing and Management of Alport Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2022, 17, 143-154. | 4.5 | 49 |
| 18 | Metabolic control analysis of the Warburg-effect in proliferating vascular smooth muscle cells. Journal of Biomedical Science, 2005, 12, 827-834. | 7.0 | 48 |

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|---|---|---|---|
| 19 | Genetics in chronic kidney disease: conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney International, 2022, 101, 1126-1141. | 5.2 | 46 |
| 20 | 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, . | 10.3 | 43 |
| 21 | Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024. | 7.9 | 43 |
| 22 | Multisystem inflammation and susceptibility to viral infections in human ZNFX1 deficiency. Journal of Allergy and Clinical Immunology, 2021, 148, 381-393. | 2.9 | 40 |
| 23 | Novel PKD1 and PKD2 mutations in autosomal dominant polycystic kidney disease (ADPKD). Nephrology Dialysis Transplantation, 2011, 26, 2181-2188. | 0.7 | 36 |
| 24 | Next generation sequencing as a useful tool in the diagnostics of mosaicism in Alport syndrome. Gene, 2013, 526, 474-477. | 2.2 | 33 |
| 25 | Exome Sequencing and Identification of Phenocopies in Patients With Clinically Presumed Hereditary Nephropathies. American Journal of Kidney Diseases, 2020, 76, 460-470. | 1.9 | 33 |
| 26 | Identification of 47 novel mutations in patients with Alport syndrome and thin basement membrane nephropathy. Pediatric Nephrology, 2016, 31, 941-955. | 1.7 | 32 |
| 27 | Pseudodominant inheritance of nephronophthisis caused by a homozygous NPHP1 deletion. Pediatric Nephrology, 2011, 26, 967-971. | 1.7 | 26 |
| 28 | Expanding the mutation spectrum for Fraser syndrome: Identification of a novel heterozygous deletion in FRAS1. Gene, 2013, 520, 194-197. | 2.2 | 24 |
| 29 | Missense mutations in EYA1 and TCF2 are a rare cause of urinary tract malformations. Nephrology Dialysis Transplantation, 2007, 23, 777-779. | 0.7 | 23 |
| 30 | Multiple urinary tract malformations with likely recessive inheritance in a large Somalian kindred. Nephrology Dialysis Transplantation, 2004, 19, 3172-3175. | 0.7 | 22 |
| 31 | 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. | 2.4 | 22 |
| 32 | 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. | 1.7 | 19 |
| 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. | 2.2 | 18 |
| 34 | Blood DNA methylation provides an accurate biomarker of KMT2B-related dystonia and predicts onset. Brain, 2022, 145, 644-654. | 7.6 | 18 |
| 35 | Mapping of a new locus for congenital anomalies of the kidney and urinary tract on chromosome 8q24. Nephrology Dialysis Transplantation, 2010, 25, 1496-1501. | 0.7 | 17 |
| 36 | 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. | 5.2 | 17 |

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|---|---|---|---|
| 37 | Heterozygous COL4A3 Variants in Histologically Diagnosed Focal Segmental Glomerulosclerosis. Frontiers in Pediatrics, 2018, 6, 171. | 1.9 | 16 |
| 38 | 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 |
| 39 | Novel heterozygous COL4A3 mutation in a family with late-onset ESRD. Pediatric Nephrology, 2010, 25, 1539-1542. | 1.7 | 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. Frontiers in Pediatrics, 2017, 5, 251. | 1.9 | 14 |
| 41 | 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 |
| 42 | Mutational analysis in 119 families with nephronophthisis. Pediatric Nephrology, 2007, 22, 366-370. | 1.7 | 13 |
| 43 | Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. European Journal of Human Genetics, 2021, 29, 411-421. | 2.8 | 13 |
| 44 | DAAM2 Variants Cause Nephrotic Syndrome via Actin Dysregulation. American Journal of Human Genetics, 2020, 107, 1113-1128. | 6.2 | 12 |
| 45 | The 2019 and 2021 International Workshops on Alport Syndrome. European Journal of Human Genetics, 2022, 30, 507-516. | 2.8 | 12 |
| 46 | Variation of the clinical spectrum and genotype-phenotype associations in Coenzyme Q10 deficiency associated glomerulopathy. Kidney International, 2022, 102, 592-603. | 5.2 | 12 |
| 47 | A De Novo Missense Variant in POU3F2 Identified in a Child with Global Developmental Delay. Neuropediatrics, 2018, 49, 401-404. | 0.6 | 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. Frontiers in Pediatrics, 2019, 7, 485. | 1.9 | 11 |
| 49 | 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. | 2.8 | 11 |
| 50 | Clinical and histological presentation of 3 siblings with mutations in the NPHP4 gene. American Journal of Kidney Diseases, 2004, 43, 358-364. | 1.9 | 10 |
| 51 | Foreign body in the bladder mimicking nephritis. Pediatric Nephrology, 2007, 22, 467-470. | 1.7 | 10 |
| 52 | Truncating <b><i>Wilms Tumor Suppressor Gene 1</i></b> Mutation in an XX Female with Adult-Onset Focal Segmental Glomerulosclerosis and Streak Ovaries: A Case Report. Nephron, 2017, 135, 72-76. | 1.8 | 10 |
| 53 | A novel interstitial deletion of 10q24.2q24.32 in a patient with renal coloboma syndrome. European Journal of Medical Genetics, 2012, 55, 211-215. | 1.3 | 9 |
| 54 | 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. | 2.2 | 9 |

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|---|---|---|---|
| 55 | Mutations in INF2 may be associated with renal histology other than focal segmental glomerulosclerosis. Pediatric Nephrology, 2018, 33, 433-437. | 1.7 | 9 |
| 56 | MAP2 â€“ A Candidate Gene for Epilepsy, Developmental Delay and Behavioral Abnormalities in a Patient With Microdeletion 2q34. Frontiers in Genetics, 2018, 9, 99. | 2.3 | 9 |
| 57 | Identification of co-occurrence in a patient with Dent's disease and ADA2-deficiency by exome sequencing. Gene, 2018, 649, 23-26. | 2.2 | 8 |
| 58 | 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. | 0.7 | 7 |
| 59 | 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. | 2.0 | 7 |
| 60 | Clonal hematopoiesis as a pitfall in germline variant interpretation in the context of Mendelian disorders. Human Molecular Genetics, 2022, 31, 2386-2395. | 2.9 | 7 |
| 61 | Haemophilus paraphrophilus, a rare cause of intracerebral abscess in children. European Journal of Pediatrics, 2008, 167, 629-632. | 2.7 | 6 |
| 62 | Congenital lymphedema as a rare and first symptom of tuberous sclerosis complex. Gene, 2020, 753, 144815. | 2.2 | 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. Gene, 2020, 742, 144542. | 2.2 | 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. Birth Defects Research, 2022, 114, 478-486. | 1.5 | 6 |
| 65 | Cyclosporine A responsive congenital nephrotic syndrome with single heterozygous variants in NPHS1, NPHS2, and PLCE1. Pediatric Nephrology, 2018, 33, 1269-1272. | 1.7 | 5 |
| 66 | Refining Kidney Survival in 383 Genetically Characterized Patients With Nephronophthisis. Kidney International Reports, 2022, 7, 2016-2028. | 0.8 | 5 |
| 67 | Disorders of sex development and Diamond-Blackfan anemia: is there an association?. Pediatric Nephrology, 2010, 25, 1255-1261. | 1.7 | 4 |
| 68 | 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. | 1.8 | 4 |
| 69 | Steroid-resistentes nephrotisches Syndrom. Medizinische Genetik, 2018, 30, 410-421. | 0.2 | 3 |
| 70 | BK virus induced nephritis in a boy with acute myeloid leukaemia undergoing bone marrow transplantation. CKJ: Clinical Kidney Journal, 2008, 1, 336-339. | 2.9 | 2 |
| 71 | 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. | 0.6 | 2 |
| 72 | Different factor H-related protein patterns in siblings with typical hemolytic uremic syndrome. Pediatric Nephrology, 2011, 26, 1345-1347. | 1.7 | 2 |

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|---|---|---|---|
| 73 | Rare co-occurrence of osteogenesis imperfecta type I and autosomal dominant polycystic kidney disease. World Journal of Pediatrics, 2016, 12, 501-503. | 1.8 | 2 |
| 74 | 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. | 0.2 | 2 |
| 75 | 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. | 2.3 | 1 |
| 76 | Exome sequencing in individuals with cardiovascular laterality defects identifies potential candidate genes. European Journal of Human Genetics, 2022, , . | 2.8 | 1 |
| 77 | No Impact of the Analytical Method Used for Determining Cystatin C on Estimating Glomerular Filtration Rate in Children. Frontiers in Pediatrics, 2017, 5, 66. | 1.9 | 0 |
| 78 | Genetische Ursachen und Therapie beim Alport-Syndrom. Medizinische Genetik, 2019, 30, 429-437. | 0.2 | 0 |