ALPART working group. Reassuring pregnancy outcomes in women with mild COL4A3-5-related disease (Alport syndrome) and genetic type of disease can aid personalized counseling.

Gosselink ME, Snoek R, Cerkauskaite-Kerpauskiene A, van Bakel SPJ, Vollenberg R, Groen H, Cerkauskiene R, Miglinas M, Attini R, Tory K, Claes KJ, van Calsteren K, Servais A, de Jong MFC, Gillion V, Vogt L, Mastrangelo A, Furlano M, Torra R, Bramham K, Wiles K, Ralston ER, Hall M, Liu L, Hladunewich MA, Lely AT, van Eerde AM;
Kidney Int. 2024 May;105(5):1088-1099. doi: 10.1016/j.kint.2024.01.034. Epub 2024 Feb 19. PMID: 38382843.

Bardet-Biedl syndrome improved diagnosis criteria and management: Inter European Reference Networks consensus statement and recommendations.

Dollfus H, Lilien MR, Maffei P, Verloes A, Muller J, Bacci GM, Cetiner M, van den Akker ELT, Grudzinska Pechhacker M, Testa F, Lacombe D, Stokman MF, Simonelli F, Gouronc A, Gavard A, van Haelst MM, Koenig J, Rossignol S, Bergmann C, Zacchia M, Leroy BP, Mosbah H, Van Eerde AM, Mekahli D, Servais A, Poitou C, Valverde D.
Eur J Hum Genet. 2024 Jul 31. doi: 10.1038/s41431-024-01634-7. Epub ahead of print. PMID: 39085583.

Complement Terminal Pathway Activation and Intrarenal Immune Response in C3 Glomerulopathy.

Marie-Sophie Meuleman, Florent Petitpre, Matthew C Pickering, Moglie Le Quintrec, Mikel Rezola Artero, Anna Duval, Marion Rabant, Alyssa Gilmore, Olivia Boyer, Julien Hogan, Aude Servais, François Provot, Vivianne Gnemmi, Maeva Eloudzeri, Anne Grunenwald, David Buob, Jean-Jacques Boffa, Anissa Moktefi, Vincent Audard, Jean-Michel Goujon, Frank Bridoux, Eric Thervet, Alexandre Karras, Lubka T Roumenina, Véronique Frémeaux Bacchi, Jean-Paul Duong Van Huyen, Sophie Chauvet
J Am Soc Nephrol. 2024 Aug 1;35(8):1034-1044. doi: 10.1681/ASN.0000000000000373. Epub 2024 May 6. PMID: 38709564 PMCID: PMC11377803

Correction: The 2019 and 2021 International workshops on Alport syndrome.

Daga S, Ding J, Deltas C, Savige J, Lipska-Ziętkiewicz BS, Hoefele J, Flinter F, Gale DP, Aksenova M, Kai H, Perin L, Barua M, Torra R, Miner JH, Massella L, Ljubanović DG, Lennon R, Weinstock AB, Knebelmann B, Cerkauskaite A, Gear S, Gross O, Turner AN, Baldassarri M, Pinto AM, Renieri A
Eur J Hum Genet. 2024 Jan;32(1):130. doi: 10.1038/s41431-023-01286-z. Erratum for:
Eur J Hum Genet. 2022 May;30(5):507-516. doi: 10.1038/s41431-022-01075-0. PMID: 36690832; PMCID: PMC10772067.

Development of clinical and laboratory biomarkers in an international cohort of 428 children with lupus nephritis.

Chiara De Mutiis, Scott E Wenderfer, Biswanath Basu, Arvind Bagga, Alvaro Orjuela, Tanmoy Sar, Amita Aggarwal, Avinash Jain, Olivia Boyer, Hui-Kim Yap, Shuichi Ito, Ai Ohnishi, Naomi Iwata , Ozgur Kasapcopur, Audrey Laurent, Eugene Yu-Hin Chan, Antonio Mastrangelo, Masao Ogura, Yuko Shima, Pornpimol Rianthavorn, Clovis A Silva, Vitor Trindade, Kjell Tullus
Multicenter Study Pediatr Nephrol. 2024 Oct;39(10):2959-2968. doi: 10.1007/s00467-024-06405-6. Epub 2024 May 28. PMID: 38802607

Diffuse Endocapillary Glomerulonephritis in a Child With IL-17RA Deficiency Emphasizes the Pivotal Role of the Complement Cascade and Anaphylatoxins.

d'Izarny-Gargas T, Grapin M, Grunenwald A, Duong-Van-Huyen JP, Rabant M, Lévy R, Puel A, Toubiana J, Boyer O, Frémeaux-Bacchi V, Charbit M, Isnard P.

Kidney Int Rep. 2023 Aug 26;8(11):2488-2491. doi: 10.1016/j.ekir.2023.08.022. eCollection 2023 Nov.PMID: 38025236

Effect of urine alkalization on urinary inflammatory markers in cystinuric patients.

Prot-Bertoye C, Jung V, Tostivint I, Roger K, Benoist JF, Jannot AS, Van Straaten A, Knebelmann B, Guerrera IC, Courbebaisse M

Clin Kidney J. 2024 Feb 22;17(3):sfae040. doi: 10.1093/ckj/sfae040. PMID: 38510798; PMCID: PMC10953617. .

Fertility Management in Cystinosis: A Clinical Perspective.

Langman CB, Delos Santos RB, Ghossein C, Atherton AM, Levtchenko EN, Servais A.

Kidney Int Rep. 2023 Nov 3;9(2):214-224. doi: 10.1016/j.ekir.2023.10.030. PMID: 38344731; PMCID: PMC10851017

Genome-wide analysis identifies MYH11 compound heterozygous variants leading to visceral myopathy corresponding to late-onset form of megacystis-microcolon-intestinal hypoperistalsis syndrome

Billon C, Piccoli GB, de Sainte Agathe JM, Stoeva R, Derive N, Heidet L, Berrebi D, Bruneval P, Jeunemaitre X, Hureaux M.

Mol Genet Genomics. 2024, 99

HDR syndrome: Large cohort and systematic review.

Lafond-Rive V, Jonard L, Loundon N, Achard S, Heidet L, Mosnier I, Lyonnet S, Brioude F, Serey Gaut M, Marlin S.

Clin Genet. 2024 106(5):564-573. doi: 10.1111/cge.14583. PMID: 38940299 Rive Le Gouard N,

HERA Clinical Trial Group. A Randomized Controlled Clinical Trial Testing Effects of Lademirsen on Kidney Function Decline in Adults with Alport Syndrome.

Gale DP, Gross O, Wang F, Esteban de la Rosa RJ, Hall M, Sayer JA, Appel G, Hariri A, Liu S, Maski M, Shen Y, Zhang Q, Iqbal S, Kowthalam MU, Lin J, Ding J

Clin J Am Soc Nephrol. 2024 Aug 1;19(8):995-1004. doi: 10.2215/CJN.0000000000000458. Epub 2024 Jun 3. PMID: 38829703; PMCID: PMC11321738.

HYDROchlorothiazide versus placebo to PROTECT polycystic kidney disease patients and improve their quality of life: study protocol and rationale for the HYDRO-PROTECT randomized controlled trial.

Bais T, Meijer E, Kramers BJ, Vart P, Vervloet M, Salih M, Bammens B, Demoulin N, Todorova P, Müller RU, Halbritter J, Paliege A, Gall EC, Knebelmann B, Torra R, Ong ACM, Karet Frankl FE, Gansevoort RT

Trials. 2024 Feb 14;25(1):120. doi: 10.1186/s13063-024-07952-x. PMID: 38355627; PMCID: PMC10865620.

IgA nephropathy in children with minimal proteinuria: to biopsy or not to biopsy?

Alexandra Cambier, Jean-Philippe Roy , Claire Dossier, Natacha Patey, Marion Rabant, Olivia Boyer, Jean Daniel Delbet, Anne-Laure Lapeyraque, Julien Hogan

Pediatr Nephrol. 2024 Mar;39(3):781-787. doi: 10.1007/s00467-023-06121-7. Epub 2023 Sep 12. PMID: 37698655

IgG-immunoadsorptions and eculizumab combination in STEC-hemolytic and uremic syndrome pediatric patients with neurological involvement.

Charlotte Duneton, Theresa Kwon, Claire Dossier, Veronique Baudouin, Marc Fila, Patricia Mariani-Kurkdijan, Isabelle Nel, Olivia Boyer, Julien Hogan

Pediatr Nephrol. 2024 Sep 19. doi: 10.1007/s00467-024-06418-1. Epub ahead of print. PMID: 39297957

IPNA clinical practice recommendations for the diagnosis and management of children with IgA nephropathy and IgA vasculitis nephritis.

Marina Vivarelli, Susan Samuel, Rosanna Coppo, Jonathan Barratt, Melvin Bonilla-Felix, Dieter Haffner, Keisha Gibson, Mark Haas, Maher Ahmed Abdel-Hafez, Marta Adragna, Paul Brogan, Siah Kim, Isaac Liu, Zhi-Hong Liu, Mukta Mantan, Yuko Shima, Masaki Shimuzu, Qian Shen, Hernan Trimarchi, Deirdre Hahn, Elisabeth Hodson, Ken Pfister, Areefa Alladin, Olivia Boyer, Koichi Nakanishi; International Pediatric Nephrology Association

Review Pediatr Nephrol. 2024 Sep 27. doi: 10.1007/s00467-024-06502-6. Online ahead of print. PMID: 39331079

Long-term outcomes of childhood-onset systemic lupus erythematosus.

A Mirguet, F A Aeschlimann, I Lemelle, R Jaussaud, P Decker, T Moulinet, S Mohamed, P Quartier, M Hofer, O Boyer, A Belot, A Hummel, N Costedoat-Chalumeau, B Bader-Meunier

Rheumatology (Oxford). 2024 Jul 15:keae344. doi: 10.1093/rheumatology/keae344. Online ahead of print. PMID: 39008948 DOI: 10.1093/rheumatology/keae344

Long-term urological and nephrological outcome after in-utero incision of obstructive duplex-system ureterocele.

Vinit N, Heidet L, Taghavi K, Salomon LJ, Ville Y, Blanc T; Collaborators

Ultrasound Obstet Gynecol. 2024, doi: 10.1002:44.

Multipopulation genome-wide association meta-analysis in pediatric steroid-sensitive nephrotic syndrome.

Boyer O, Dorval G

Kidney Int. 2024 Jan;105(1):14-17. doi: 10.1016/j.kint.2023.08.022. Epub 2023 Sep 13.PMID: 37714428

Performance and clinical utility of a new supervised machine-learning pipeline in detecting rare ciliopathy patients based on deep phenotyping from electronic health records and semantic similarity.

Carole Faviez, Marc Vincent, Nicolas Garcelon, Olivia Boyer, Bertrand Knebelmann, Laurence Heidet, Sophie Saunier, Xiaoyi Chen, Anita Burgun

Orphanet J Rare Dis. 2024 Feb 10;19(1):55. doi: 10.1186/s13023-024-03063-7. PMID: 38336713 PMCID: PMC10858490

Prevalence of Fabry Disease in Patients on Dialysis in France.

Sens F, Guittard L, Knebelmann B, Moranne O, Choukroun G, de Précigout V, Couchoud C, Deleruyelle I, Lancelot L, Tran Thi Phuong L, Ghafari T, Fabrydial Study Group, Juillard L, Germain DP.

Int J Mol Sci. 2024 Sep 20;25(18):10104. doi: 10.3390/ijms251810104. PMID: 39337589; PMCID: PMC11432483.

Primary hyperoxaluria in adults and children: a nationwide cohort highlights a persistent diagnostic delay.

Pszczolinski R, Acquaviva C, Berrahal I, Biebuyck N, Burtey S, Clabault K, Dossier C, Guillet M, Hemery F, Letavernier E, Rousset-Rouvière C, Bacchetta J, Moulin B.

Clin Kidney J. 2024 Apr 3;17(5):sfae099. doi: 10.1093/ckj/sfae099. eCollection 2024 May.PMID: 38737343

Recent Developments in the Treatment of Pediatric Distal Renal Tubular Acidosis.

Boyer O, Ould Rabah M, Preka E.

Paediatr Drugs. 2024 Sep 26. doi: 10.1007/s40272-024-00651-9. Epub ahead of print. PMID: 39325135.

Recombinant ADAMTS13 in Congenital Thrombotic Thrombocytopenic Purpura.

Scully M, Antun A, Cataland SR, Coppo P, Dossier C, Biebuyck N, Hassenpflug WA, Kentouche K, Knöbl P, Kremer Hovinga JA, López-Fernández MF, Matsumoto M, Ortel TL, Windyga J, Bhattacharya I, Cronin M, Li H, Mellgård B, Patel M, Patwari P, Xiao S, Zhang P, Wang LT;

cTTP Phase 3 Study Investigators. N Engl J Med. 2024 May 2;390(17):1584-1596. doi: 10.1056/NEJMoa2314793.PMID: 38692292

Renal and Extrarenal Phenotypes in Patients With HNF1B Variants and Chromosome 17q12 Microdeletions HNF1B variant study group.

Buffin-Meyer B, Richard J, Guigonis V, Weber S, König J, Heidet L, Moussaoui N, Vu JP, Faguer S, Casemayou A, Prakash R, Baudouin V, Hogan J, Alexandrou D, Bockenhauer D, Bacchetta J, Ranchin B, Pruhova S, Zieg J, Lahoche A, Okorn C, Antal-Kónya V, Morin D, Becherucci F, Habbig S, Liebau MC, Mauras M, Nijenhuis T, Llanas B, Mekahli D, Thumfart J, Tönshoff B, Massella L, Eckart P, Cloarec S, Cruz A, Patzer L, Roussey G, Vrillon I, Dunand O, Bessenay L, Taroni F, Zaniew M, Louillet F, Bergmann C, Schaefer F, van Eerde AM, Schanstra JP, Decramer S

Kidney Int Rep. 2024 9:2514-2526. doi: 10.1016/j.ekir.2024.05.007. eCollection 2024 Aug. PMID: 39156164

Single-cell transcriptomics identifies aberrant glomerular angiogenic signalling in the early stages of WT1 kidney disease.

Jennifer C Chandler, Daniyal J Jafree, Saif Malik, Gideon Pomeranz , Mary Ball, Maria Kolatsi-Joannou, Alice Piapi , William J Mason, Andrew V Benest, David O Bates, Aleksandra Letunovska, Reem Al-Saadi, Marion Rabant, Olivia Boyer, Kathy Pritchard-Jones, Paul J Winyard, Andrew S Mason, Adrian S Woolf, Aoife M Waters, David A Long

J Pathol. 2024 Oct;264(2):212-227. doi: 10.1002/path.6339. Epub 2024 Aug 23. PMID: 39177649

Social Deprivation and Incidence of Pediatric Kidney Failure in France.

Bénédicte Driollet, Cécile Couchoud, Justine Bacchetta, Olivia Boyer, Julien Hogan, Denis Morin, François Nobili, Michel Tsimaratos, Etienne Bérard, Florian Bayer, Ludivine Launay, Karen Leffondré, Jérôme Harambat

Kidney Int Rep. 2024 Apr 26;9(7):2269-2277. doi: 10.1016/j.ekir.2024.04.042. eCollection 2024 Jul. PMID: 39081742 PMCID: PMC11284436

Steroid-Resistant Nephrotic Syndrome due to NPHS2 Variants Is Not Associated With Posttransplant Recurrence.

Kachmar J, Boyer O, Lipska-Ziętkiewicz B, Morinière V, Gribouval O, Heidet L, Balasz-Chmielewska I, Benetti E, Cloarec S, Csaicsich D, Decramer S, Gellermann J, Guigonis V, Hogan J, Bayazit AK, Melk A, Nigmatullina N, Oh J, Ozaltin F, Ranchin B, Tsimaratos M, Trautmann A, Antignac C, Schaefer F, Dorval G; PodoNet Network

Kidney Int Rep. 2024, 9:973-981. doi: 10.1016

.

Targeted RNAseq from patients' urinary cells to validate pathogenic noncoding variants in autosomal dominant polycystic kidney disease genes: a proof of concept.

Dorval G, Le Gac G, Morinière V, Ka C, Goursaud C, Knebelmann B, Marijon P, Nambot S, Cagnard N, Nitschké P, Michel-Calemard L, Audrézet MP, Heidet L.

Kidney Int. 2024 Sep;106(3):532-535. doi: 10.1016/j.kint.2024.05.029. Epub 2024 Jun 27. PMID: 38944240.

To biopsy or not to biopsy a teenager with typical idiopathic nephrotic syndrome? Start steroids first.

Boyer O, Bernardi S, Preka E

Pediatr Nephrol. 2024 Sep 11. doi: 10.1007/s00467-024-06447-w. Online ahead of print.PMID: 39259322

Unusual familial cystic kidney disease: combining fine radiologic and genetic evaluation to solve the case.

Bodard AS, Nabbout R, Hélénon O, Knebelmann B.

BMC Nephrol. 2024 Sep 30;25(1):325. doi: 10.1186/s12882-024-03747-z. PMID: 39350077; PMCID: PMC11443641.

Vesico-ureteral reflux diagnosis after initial kidney abscess: Results from a Paediatric Tertiary Hospital.

Preka E, Miller N, Avramescu M, Berteloot L, Vinit N, Botto N, Grapin M, Prévot M, Boistault M, Garcelon N, Taghavi K, Schrimpf C, Cohen JF, Blanc T, Boyer O.

Acta Paediatr. 2024 Jul 5. doi: 10.1111/apa.17353. Epub ahead of print. PMID: 38967007.

Voice of a caregiver: call for action for multidisciplinary teams in the care for children with atypical hemolytic uremic syndrome.

Linda Burke, Sidharth Kumar Sethi, Olivia Boyer, Christoph Licht, Mignon McCulloch, Raghav Shah, Valerie A Luyckx, Rupesh Raina

Editorial. Pediatr Nephrol. 2024 Jul;39(7):1961-1963. doi: 10.1007/s00467-023-06158-8. PMID: 37782345 DOI: 10.1007/s00467-023-06158-8

Worldwide disparities in access to treatment and investigations for nephropathic cystinosis: a 2023 perspective.

Regnier M, Flammier S, Boutaba M, Ndongo AA, Servais A, Schaefer F, Levtchenko E, Bacchetta J, Bertholet-Thomas A.

Pediatr Nephrol. 2024 Apr;39(4):1113-1123. doi: 10.1007/s00467-023-06179-3. Epub 2023 Nov 18. PMID: 37978055; PMCID: PMC10899370.

X-linked transient antenatal Bartter syndrome related to MAGED2 gene: enriching the phenotypic description and pathophysiologic investigation.

Buffet A, Filser M, Bruel A, Dard R, Quibel T, Dubucs C, Kwon T, Le Tanno P, Thevenon J, Ziegler A, Allard L, Guigonis V, Roux JJ, Heidet L, Rougeulle C, Boyer O, Vargas-Poussou R, Hureaux M.

Genet Med. 2024 Jul 18:101217. doi: 10.1016/j.gim.2024.101217. Online ahead of print. PMID: 39036894