EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

Curriculum Vitae

Personal information

Detlef Bockenhauer

Work experience

Dates	Detail of position held	Institution
1991-1994	Arzt im Praktikum/Assistenzarzt	Altonaer Kinder
1994-1997	Intern and Resident, Pediatrics	NYU/Bellevue,
1997-2000	Fellow in Pediatric Nephrology	Yale University
2000-2003	Associate Research Scientist	Yale University
2003-2004	Assistant Professor	Yale University
2004-2010	Consultant Nephrologist/ Honorary Senior Lecturer	Great Ormond S Institute of Chil
2011-2012	Clinical Senior Lecturer/ Honorary Consultant	Institute of Chil Great Ormond S
2012-2015	Reader/ Honorary Consultant	Institute of Chil
2014-2023	Honorary Consultant	Royal Free Hosp
2015-2023	Professor/ Honorary Consultant	UCL Departmer Great Ormond S
2019-2023	Clinical lead, renal unit	Great Ormond S
2023-	Head, Paediatric Nephrology/ Professor	UZ Leuven, D Leuven

Education and training

Dates	Detail of degree; diploma; other qualification	Institution
1982	Abitur (A-level)	Sachsenwaldgymnasium
1985-91	Medical studies	Reinbek
1991	Staatsexamen Medizin	Universität Hamburg /
1993	Approbation (registration	Lübeck
1994	as doctor)	Med. Uni. Lübeck
1997	Dr. med. (PhD)	Dept. of Health, Germany
2000	Diplomat of the American Board of	Med. Uni. Lübeck American Board of Pediatrics
2003	Pediatrics	American Board of Pediatrics
2003	Diplomat of the	
2007-	American Board of	GMC
2025	Pediatrics, Subboard Nephrology	Ärztekammer Hamburg Royal College of Paediatrics
	Specialist Registry (Paediatrics)	and Child Health
	Facharzt für Kinderheilkunde	
	Fellow	

Additional information

Publications

Publication list

Books

1. Rees L, **Bockenhauer D**, Brogan PA and Webb NJA: Paediatric Nephrology, 2nd edition, Oxford Specialist Handbooks in Paediatrics, Oxford, 2012
2. Rees L, **Bockenhauer D**, Webb NJA and Punaro M:: Paediatric Nephrology, 23d edition, Oxford Specialist Handbooks in Paediatrics, Oxford, 2017

Chapters in books

3. Lopez-Garcia SC, Walsh, SB and **Bockenhauer D**: Renal Tubular Acidosis in: Pediatric Nephrology, 7th edition, in press
4. **Bockenhauer D** and Hayes W: Disorders of Fluid and Electrolyte Balance in: Pediatric Endocrinology, in press
5. **Bockenhauer D**: Fluid, Electrolyte and Acid-Base Disorders in Children in: Brenner and Rector: The Kidney, 11th edition, in press
6. Kleta R and **Bockenhauer D**: Proximal Tubular acidosis in Lifton, R, Somlo S, Giebisch G, Pollak M and Seldin D: Genetic diseases of the kidney, 2nd edition, Elsevier, in press
7. Bichet D and **Bockenhauer D**: Nephrogenic Diabetes Insipidus in Lifton, R, Somlo S, Giebisch G, Pollak M and Seldin D: Genetic diseases of the kidney, 2nd edition, Elsevier, in press
8. **Bockenhauer D and Bichet DG**: Diabetes Insipidus in: Geary DF, Schaefer F eds. Pediatric Kidney Disease, 2nd edition, Heidelberg, Springer, in press
9. **Bockenhauer D** and Kleta, R.: Renal Fanconi syndrome and other proximal tubulopathies in: Geary DF, Schaefer F eds. Pediatric Kidney Disease, 2nd edition, Heidelberg, Springer, in press
10. Todd, A and **Bockenhauer D**: Renal Tubular Acidosis in: Geary DF, Schaefer F eds. Pediatric Kidney Disease, 2nd edition, Heidelberg, Springer, in press
11. **Bockenhauer D**: Diabetes Insipidus in: Geary DF, Schaefer F eds. Comprehensive Pediatric Nephrology, 1st edition, Philadelphia, Mosby Elsevier, 2008: 489-98
12. **Bockenhauer D:** Tubulopathies in: Clinical Pediatric Nephrology: Third Edition. 819-837
13. Van't Hoff W and **Bockenhauer D**: Fanconi Syndrome in: Geary DF, Schaefer F eds. Comprehensive Pediatric Nephrology, 1st edition, Philadelphia, Mosby Elsevier, 2008: 433-50
14. **Bockenhauer D**, Bichet DG and Unwin R: Nephrogenic Diabetes Insipidus: Oxford Desk Reference Nephrology 1st edition, Oxford, Oxford University Press, 2009
15. **Bockenhauer D**, Bichet D and Unwin R: Isolated defects of tubular function: Oxford Desk Reference Nephrology 1st edition, Oxford, Oxford University Press, 2009
16. **Bockenhauer D**: Primary Hyperoxalurias: Oxford Desk Reference Nephrology 1st edition, Oxford, Oxford University Press, 2009
17. **Bockenhauer D** and Kleta R: Approach to the patient with salt-wasting states: Oxford Textbook of Clinical Nephrology, 4th edition, 2015
18. Kleta R and **Bockenhauer D**: Approach to the patient with Fanconi syndrome, glycosuria and aminoaciduria: Oxford Textbook of Clinical Nephrology, 4th edition, 2015
19. Satlin L and **Bockenhauer D**: Physiology of the Developing Kidney: Potassium Homeostasis and its Disorder in Pediatric Nephrology, edited by Ellis D. Avner, William E. Harmon, Patrick Niaudet, Norishige Yoshikawa, Francesco Emma and Stuart L. Goldstein. 2015

20. **Bockenhauer D**: Inherited disorders of salt and water, Comprehensive Clinical Nephrology, 6th edition, in press

21. Besouw M and **Bockenhauer D**: Potassium Metabolism in: Nephrology and Fluid/Electrolyte Physiology, 3rd edition, in press

22. Hayes, W., & Bockenhauer, D. (2024). Disorders of Fluid and Electrolyte Balance. In *Endocrinology (Switzerland)* (Vol. Part F3598, pp. 373-390). doi:10.1007/978-3-030-23709-7_9

Journal Articles

Original publications

1. Haffner, D., Emma, F., Seefried, L., Högler, W., Javaid, K. M., Bockenhauer, D., . . . Linglart, A. (2025). Clinical practice recommendations for the diagnosis and management of X-linked hypophosphataemia.. *Nat Rev Nephrol*. doi:10.1038/s41581-024-00926-x
2. Bökenkamp, A., Ariceta, G., Böckenhauer, D., Devuyst, O., Emma, F., van Bennekom, D., . . . Prikhodina, L. (2025). Dent disease: Clinical Practice Recommendations.. *Nephrol Dial Transplant*. doi:10.1093/ndt/gfaf003
3. Tan, H. L., Marlais, M., Veligratli, F., Shah, S., Hayes, W., & Bockenhauer, D. (2024). Real world experience with potassium citrate and potassium hydrogen carbonate prolonged release granules (Sibnayal) in paediatric renal tubular acidosis. In *PEDIATRIC NEPHROLOGY* Vol. 39 (pp. S270). SPRINGER.
4. Sadeghi-Alavijeh, O., Chan, M. M., Doctor, G. T., Voinescu, C. D., Stuckey, A., Kousathanas, A., . . . Gale, D. P. (2024). Quantifying variant contributions in cystic kidney disease using national-scale whole genome sequencing. *The Journal of Clinical Investigation (JCI)*. doi:10.1172/JCI181467
5. Buffin-Meyer, B., Richard, J., Guigonis, V., Weber, S., Koenig, J., Heidet, L., . . . Decramer, S. (2024). Renal and Extrarenal Phenotypes in Patients With HNF1B Variants and Chromosome 17q12 Microdeletions. *KIDNEY INTERNATIONAL REPORTS*, *9*(8), 2514-2526. doi:10.1016/j.ekir.2024.05.007
6. Wong, K., Pitcher, D., Braddon, F., Downward, L., Steenkamp, R., Masoud, S., . . . Gale, D. P. (2024). Description and Cross-Sectional Analyses of 25,880 Adults and Children in the UK National Registry of Rare Kidney Diseases Cohort. *Kidney International Reports*, *9*(7), 2067-2083. doi:10.1016/j.ekir.2024.04.062
7. Tan, H. L., Marlais, M., Veligratli, F., Shah, S., Hayes, W., & Bockenhauer, D. (2024). Treatment of paediatric renal tubular acidosis with a prolonged-release alkali supplementation. *Pediatric Nephrology*. doi:10.1007/s00467-024-06411-8
8. Verjans, M., Hindryckx, A., Rosier, K., Devriendt, K., Mekahli, D., & Bockenhauer, D. (2024). Antenatal presentation and early postnatal treatment of infantile hypercalcemia type 2.. *Pediatr Nephrol*. doi:10.1007/s00467-024-06403-8
9. Voinescu, C. D., Mozere, M., Genovese, G., Downie, M. L., Gupta, S., Gale, D. P., . . . Stanescu, H. C. (2024). A Neanderthal haplotype introgressed into the human genome confers protection against membranous nephropathy. *Kidney International*. doi:10.1016/j.kint.2024.01.017
10. Sadeghi-Alavijeh, O., Chan, M. M. Y., Doctor, G., Voinescu, C., Stuckey, A., Kousathanas, A., . . . Gale, D. (2024). Quantifying variant contributions in cystic kidney disease using national-scale whole genome sequencing. doi:10.1101/2024.02.14.24302377
11. Levtchenko, E., Ariceta, G., Arguedas Flores, O., Bichet, D. G., Bockenhauer, D., Emma, F., . . . Knoers, N. V. A. M. (n.d.). International expert consensus statement on the diagnosis and management of congenital nephrogenic diabetes insipidus (arginine vasopressin resistance). *NATURE REVIEWS NEPHROLOGY*, 14 pages. doi:10.1038/s41581-024-00897-z
12. Zieg, J., Thomasova, D., Libik, M., Sumnik, Z., & Bockenhauer, D. (n.d.). Hyperkalaemic acidosis: blood pressure is the diagnostic clue. *PEDIATRIC NEPHROLOGY*, 4 pages. doi:10.1007/s00467-024-06590-4
13. Wong, K., Pitcher, D., Braddon, F., Downward, L., Steenkamp, R., Annear, N., . . . Gale, D. P. (2024). Effects of rare kidney diseases on kidney failure: a longitudinal analysis of the UK National Registry of Rare Kidney Diseases (RaDaR) cohort. *The Lancet*, *403*(10433), 1279-1289. doi:10.1016/S0140-6736(23)02843-X
14. Wu, J. G., Guha, C., Hughes, A., Torrisi, L. G., Craig, J. C., Sinha, A., . . . Jaure, A. (2025). Patient, Parental, and Health Professional Perspectives on Growth in Children With CKD. *American Journal of Kidney Diseases*, *85*(1), 14-24.e1. doi:10.1053/j.ajkd.2024.06.016 Walker, E. Y. X., Lindsay, T. A. J., Allgrove, J., Marlais, M., Bockenhauer, D., & Hayes, W. (2023). Burosumab in management of X-linked hypophosphataemia: a retrospective cohort study of growth and serum phosphate levels.. *Arch Dis Child*. doi:10.1136/archdischild-2022-324962
15. Kiparissi, F., Dastamani, A., Palm, L., Azabdaftari, A., Campos, L., Gaynor, E., . . . Jones, K. D. J. (2023). Phosphomannomutase 2 (PMM2) variants leading to hyperinsulinism-polycystic kidney disease are associated with early-onset inflammatory bowel disease and gastric antral foveolar hyperplasia. *Human Genetics*. doi:10.1007/s00439-023-02523-7
16. Wan, E. R., Iancu, D., Ashton, E., Siew, K., Mohidin, B., Sung, C. C., . . . Walsh, S. B. (2022). Machine Learning to Identify Genetic Salt-Losing Tubulopathies in Hypokalemic Patients. *Kidney International Reports*. doi:10.1016/j.ekir.2022.12.008
17. Smeulders, N., Cho, A., Alshaiban, A., Read, K., Fagan, A., Easty, M., . . . Bockenhauer, D. (2022). Shockwaves and the Rolling Stones: An Overview of Pediatric Stone Disease. *Kidney International Reports*. doi:10.1016/j.ekir.2022.11.017
18. Verploegen, M. F. A., Vargas-Poussou, R., Walsh, S. B., Alpay, H., Amouzegar, A., Ariceta, G., . . . Nijenhuis, T. (2022). Parathyroid hormone and phosphate homeostasis in patients with Bartter and Gitelman syndrome: an international cross-sectional study.. *Nephrology Dialysis Transplantation*. doi:10.1093/ndt/gfac029
19. Burballa, C., Cantero-Recasens, G., Prikhodina, L., Lugani, F., Schlingmann, K., Ananin, P. V., . . . DENT study group. (2022). Clinical and genetic characteristics of Dent's Disease type 1 in Europe.. *Nephrol Dial Transplant*. doi:10.1093/ndt/gfac310
20. Downie, M. L., Gupta, S., Chan, M. M. Y., Sadeghi-Alavijeh, O., Cao, J., Parekh, R. S., . . . Gale, D. P. (2022). Shared genetic risk across different presentations of gene test–negative idiopathic nephrotic syndrome. *Pediatric Nephrology*. doi:10.1007/s00467-022-05789-7
21. Veys, K., Zadora, W., Hohenfellner, K., Bockenhauer, D., Janssen, M. C. H., Niaudet, P., . . . Levtchenko, E. (2022). Outcome of infantile nephropathic cystinosis depends on early intervention, not genotype: a multicenter sibling cohort study.. *Journal of Inherited Metabolic Disease*. doi:10.1002/jimd.12562
22. Sinha R, Pradhan S, Banerjee S, Jahan A, Akhtar S, Pahari A, Raut S, Parakh P, Basu S, Srivastava P, Nayak S, Thenral SG, Ramprasad V, Ashton E, Bockenhauer D, Mandal K.: Whole-exome sequencing and variant spectrum in children with suspected inherited renal tubular disorder: the East India Tubulopathy Gene Study. Pediatr Nephrol. 2022 Jan 10. doi: 10.1007/s00467-021-05388-y. Online ahead of print
23. Patel V, Klootwijk E, Whiting G, Bockenhauer D, Siew K, Walsh S, Bleich M, Himmerkus N, Jaureguiberry G, Issler N, Godovac-Zimmermann J, Kleta R, Wheeler J.: Quantification of FAM20A in human milk and identification of calcium metabolism proteins. Physiol Rep. 2021 Dec;9(24):e15150. doi: 10.14814/phy2.15150.
24. 100,000 Genomes Project Pilot Investigators, Smedley D, Smith KR, Martin A, Thomas EA, McDonagh EM,

Cipriani V, Ellingford JM, Arno G, Tucci A, Vandrovcova J, Chan G, Williams HJ, Ratnaike T, Wei W, Stirrups K, Ibanez K, Moutsianas L, Wielscher M, Need A, Barnes MR, Vestito L, Buchanan J, Wordsworth S, Ashford S, Rehmström K, Li E, Fuller G, Twiss P, Spasic-Boskovic O, Halsall S, Floto RA, Poole K, Wagner A, Mehta SG, Gurnell M, Burrows N, James R, Penkett C, Dewhurst E, Gräf S, Mapeta R, Kasanicki M, Haworth A, Savage H, Babcock M, Reese MG, Bale M, Baple E, Boustred C, Brittain H, de Burca A, Bleda M, Devereau A, Halai D, Haraldsdottir E, Hyder Z, Kasperaviciute D, Patch C, Polychronopoulos D, Matchan A, Sultana R, Ryten M, Tavares ALT, Tregidgo C, Turnbull C, Welland M, Wood S, Snow C, Williams E, Leigh S, Foulger RE, Daugherty LC, Niblock O, Leong IUS, Wright CF, Davies J, Crichton C, Welch J, Woods K, Abulhoul L, Aurora P, Bockenhauer D, Broomfield A, Cleary MA, Lam T, Dattani M, Footitt E, Ganesan V, Grunewald S, Compeyrot-Lacassagne S, Muntoni F, Pilkington C, Quinlivan R, Thapar N, Wallis C, Wedderburn LR, Worth A, Bueser T, Compton C, Deshpande C, Fassihi H, Haque E, Izatt L, Josifova D, Mohammed S, Robert L, Rose S, Ruddy D, Sarkany R, Say G, Shaw AC, Wolejko A, Habib B, Burns G, Hunter S, Grocock RJ, Humphray SJ, Robinson PN, Haendel M, Simpson MA, Banka S, Clayton-Smith J, Douzgou S, Hall G, Thomas HB, O'Keefe RT, Michaelides M, Moore AT, Malka S, Pontikos N, Browning AC, Straub V, Gorman GS, Horvath R, Quinton R, Schaefer AM, Yu-Wai-Man P, Turnbull DM, McFarland R, Taylor RW, O'Connor E, Yip J, Newland K, Morris HR, Polke J, Wood NW, Campbell C, Camps C, Gibson K, Koelling N, Lester T, Németh AH, Palles C, Patel S, Roy NBA, Sen A, Taylor J, Cacheiro P, Jacobsen JO, Seaby EG, Davison V, Chitty L, Douglas A, Naresh K, McMullan D, Ellard S, Temple IK, Mumford AD, Wilson G, Beales P, Bitner-Glindzicz M, Black G, Bradley JR, Brennan P, Burn J, Chinnery PF, Elliott P, Flinter F, Houlden H, Irving M, Newman W, Rahman S, Sayer JA, Taylor JC, Webster AR, Wilkie AOM, Ouwehand WH, Raymond FL, Chisholm J, Hill S, Bentley D, Scott RH, Fowler T, Rendon A, Caulfield M.: 100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. N Engl J Med. 2021 Nov 11;385(20):1868-1880. doi: 10.1056/NEJMoa2035790

25. Falcone MP, Pritchard-Jones K, Brok J, Mifsud W, Williams RD, Nakata K, Tugnait S, Al-Saadi R, Side L, Anderson J, Duncan C, Marks SD, Bockenhauer D, Chowdhury T.: Long-term kidney function in children with Wilms tumour and constitutional WT1 pathogenic variant. Pediatr Nephrol. 2021 Oct 4. doi: 10.1007/s00467-021-05125-5
26. Viering D, Schlingmann KP, Hureaux M, Nijenhuis T, Mallett A, Chan MMY, van Beek A, van Eerde AM, Coulibaly JM, Vallet M, Decramer S, Pelletier S, Klaus G, Kömhoff M, Beetz R, Patel C, Shenoy M, Steenbergen EJ, Anderson G, Bongers EMHF, Bergmann C, Panneman D, Rodenburg RJ, Kleta R, Houillier P, Konrad M, Vargas-Poussou R, Knoers NVAM, Bockenhauer D, de Baaij JHF; Genomics England Research Consortium.: Gitelman-Like Syndrome Caused by Pathogenic Variants in mtDNA. J Am Soc Nephrol. 2021 Oct 4:ASN.2021050596. doi: 10.1681/ASN.2021050596
27. Emma F, Van't Hoff W, Hofenfellner K, Topaloglu R, Greco M, Ariceta G, Bettini C, Bockenhauer D, Veys K, Pape L, et al. Long-term follow-up of nephropathic cystinosis: a cohort study spanning five decades Kidney International, S0085-2538(21)00648-7. doi: 10.1016/j.kint.2021.06.019.
28. Wu JG-A, Tong A, Evangelidis N, Manera KE, Hanson CS, Baumgart A, Amir N, Sinha A, Dart A, Eddy AA, et al. Patient and caregiver perspectives on blood pressure in children with chronic kidney disease Nephrology Dialysis Transplantation 27 Aug 2021
29. Bianic F, Guelfucci F, Robin L, Martre C, Game D, Bockenhauer D. Epidemiology of Distal Renal Tubular Acidosis: A Study Using Linked UK Primary Care and Hospital Data NEPHRON 10 pages 01 Jul 2021
30. Bassanese G, Wlodkowski T, Servais A, Heidet L, Roccatello D, Emma F, Levtchenko E, Ariceta G, Bacchetta J, Capasso G, et al. The European Rare Kidney Disease Registry (ERKReg): objectives, design and initial results ORPHANET JOURNAL OF RARE DISEASES 16(1):15 pages Article number ARTN 251 02 Jun 2021
31. Boyer O, Schaefer F, Haffner D, Bockenhauer D, Holtta T, Berody S, Webb H, Heselden M, Lipska-Zietkiewicz BS, Ozaltin F, et al. Management of congenital nephrotic syndrome: consensus recommendations of the ERKNet-ESPN Working Group (vol 17, pg 277, 2021) NATURE REVIEWS NEPHROLOGY 17(6):434-434 30 Apr 2021
32. Trepiccione F, Walsh SB, Ariceta G, Boyer O, Emma F, Camilla R, Ferraro PM, Haffner D, Konrad M, Levtchenko E, et al. Distal Renal Tubular Acidosis: ERKNet/ESPN Clinical Practice Points. Nephrol Dial Transplant 29 Apr 2021
33. Islam S, Tekman M, Flanagan SE, Guay-Woodford L, Hussain K, Ellard S, Kleta R, Bockenhauer D, Stanescu H, Iancu D. Founder mutation in the PMM2 promotor causes hyperinsulinemic hypoglycaemia/polycystic kidney disease (HIPKD) Molecular Genetics and Genomic Medicine 03 Apr 2021
34. Schlingmann KP, Renigunta A, Hoorn EJ, Forst A-L, Renigunta V, Atanasov V, Mahendran S, Barakat TS, Gillion V, Godefroid N, et al. Defects in KCNJ16 Cause a Novel Tubulopathy with Hypokalemia, Salt Wasting, Disturbed Acid-Base Homeostasis, and Sensorineural Deafness. J Am Soc Nephrol 02 Apr 2021
35. Downie ML, Gupta S, Tekman MC, Cheshire C, Arora S, Licht C, Robinson LA, Munoz M, Aris AM, Al Attrach I, et al. Identification of a Locus on the X Chromosome Linked to Familial Membranous Nephropathy Kidney International Reports 03 Mar 2021
36. Chan, M. M. Y., Sadeghi-Alavijeh, O., Lopes, F. M., Hilger, A. C., Stanescu, H. C., Voinescu, C. D., **Bockenhauer, D** . . . Gale, D. P. (2022). Diverse ancestry whole-genome sequencing association study identifies TBX5 and PTK7 as susceptibility genes for posterior urethral valves. *eLife*, *11*. doi:10.7554/eLife.74777
37. Konrad M, Nijenhuis T, Ariceta G, Bertholet-Thomas A, Calo LA, Capasso G, Emma F, Schlingmann KP, Singh M, Trepiccione F, et al. Diagnosis and management of Bartter syndrome: executive summary of the consensus and recommendations from the European Rare Kidney Disease Reference Network Working Group for Tubular Disorders Kidney International 99(2):324-335 01 Feb 2021
38. Boyer O, Schaefer F, Haffner D, Bockenhauer D, Hölttä T, Bérody S, Webb H, Heselden M, Lipska-Zie Tkiewicz BS, Ozaltin F, et al. Management of congenital nephrotic syndrome: consensus recommendations of the ERKNet-ESPN Working Group. Nat Rev Nephrol 29 Jan 2021
39. Lopez-Garcia, S. C., Downie, M. L., Kim, J. S., Boyer, O., Walsh, S. B., Nijenhuis, T., . . . Bockenhauer, D. (2020). Treatment and long-term outcome in primary nephrogenic diabetes insipidus. *Nephrology Dialysis Transplantation*

40. Chromek, M., Jungner, Å., Rudolfson, N., Ley, D., Bockenhauer, D., & Hagander, L. (2020). Hyponatraemia despite isotonic maintenance fluid therapy: a time series intervention study.. *Arch Dis Child*. doi:10.1136/archdischild-2019-318555

41. Hanson, C. S., Craig, J. C., Logeman, C., Sinha, A., Dart, A., Eddy, A. A., . . . Tong, A. (2020). Establishing core outcome domains in pediatric kidney disease: report of the Standardized Outcomes in Nephrology – Children and Adolescents (SONG-KIDS) consensus workshops *Kidney International*.

42. Kari, J. A., Alhasan, K. A., Albanna, A. S., Safder, O. Y., Shalaby, M., Bockenhauer, D., & El-Desoky, S. M. (2020). Rituximab versus cyclophosphamide as first steroid sparing agent in childhood frequently relapsing and steroid dependent nephrotic syndrome. *Pediatric Nephrology*

43. Logeman, C., Guha, C., Howell, M., Hanson, C. S., Craig, J. C., Samuel, S., . . . Gutman, T. (2020). Developing consensus-based outcome domains for trials in children and adolescents with chronic kidney disease: an international Delphi survey. *American Journal of Kidney Diseases*

44. Turro, E., Astle, W. J., Megy, K., Gräf, S., Greene, D., Shamardina, O.,. . . Buckland, M. S. (2020). Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*. doi:10.1038/s41586-020-2434-2

45. Balogh, E., Chandler, J., Varga, M., Tahoun, M., Menyhárd, D. K., Schay, G., **Bockenhauer D**. . . Tory, K. (2020). Pseudouridylation defect due to DKC1 and NOP10 mutations causes nephrotic syndrome with cataracts, hearing impairment, and enterocolitis. *Proceedings of the National Academy of Sciences of the United States of America*. doi:10.1073/pnas.2002328117

46. Viering, D. H. H. M., Chan, M. M. Y., Hoogenboom, L., Iancu, D., de Baaij, J. H. F., Tullus, K., . **Bockenhauer, D**. (2020). Genetics of renovascular hypertension in children. *Journal of Hypertension*. doi:10.1097/HJH.0000000000002491

47. Giri, D., **Bockenhauer, D**., Deshpande, C., Achermann, J. C., Taylor, N. F., Rumsby, G., . . . Ajzensztejn, M. (2020). Co-Existence of Congenital Adrenal Hyperplasia and Bartter Syndrome due to Maternal Uniparental Isodisomy of HSD3B2 and CLCNKB Mutations.. *Horm Res Paediatr*, *93*(1), 66-71. doi:10.1159/000507577

48. Jdiaa, S. S., Walsh, S. B., **Bockenhauer, D**., Fakhredine, S. W., & Koubar, S. H. (2020). The hypokalemia mystery: distinguishing Gitelman and Bartter syndromes from 'pseudo-Bartter syndrome'. *Nephrol Dial Transplant*. doi:10.1093/ndt/gfaa100

49. Lipska-Ziętkiewicz, B. S., Ozaltin, F., Hölttä, T., Bockenhauer, D., Bérody, S., Levtchenko, E., . . . Boyer, O. (2020). Genetic aspects of congenital nephrotic syndrome: a consensus statement from the ERKNet–ESPN inherited glomerulopathy working group. *European Journal of Human Genetics*. doi:10.1038/s41431-020-0642-8

50. Xie, J., Liu, L., Mladkova, N., Li, Y., Ren, H., Wang, W., **Bockenhauer, D,** . . . Kiryluk, K. (2020). The genetic architecture of membranous nephropathy and its potential to improve non-invasive diagnosis.. *Nat Commun*, *11*(1), 1600. doi:10.1038/s41467-020-15383-w

51. Levey, A. S., Eckardt, K. -U., Dorman, N. M., Christiansen, S. L., Hoorn, E. J., Ingelfinger, J. R., **Bockenhauer, D** . . . Winkelmayer, W. C. (2020). Nomenclature for kidney function and disease: report of a Kidney Disease: Improving Global Outcomes (KDIGO) Consensus Conference. *Kidney International*. doi:10.1016/j.kint.2020.02.010

52. Hureaux, M., Ashton, E., Dahan, K., Houillier, P., Blanchard, A., Cormier, C., . . . **Bockenhauer, D,** Vargas-Poussou, R. (2019). High-throughput sequencing contributes to the diagnosis of tubulopathies and familial hypercalcemia hypocalciuria in adults.. *Kidney Int*. doi:10.1016/j.kint.2019.08.027

53. Malakasioti, G., Alders, N., Lucchini, G., Cheng, I. L., & **Bockenhauer, D**. (2019). Acute kidney injury in an infant with severe combined immunodeficiency: Questions. *Pediatric Nephrology*. doi:10.1007/s00467-019-04302-x

54. Tahoun, M., Chandler, J. C., Ashton, E., Haston, S., Hannan, A., Kim, J. S., **Bockenhauer, D**. . . Waters, A. M. (2019). 'Mutations in LAMB2 associate with albuminuria and Optic Nerve Hypoplasia with Hypopituitarism'.. *J Clin Endocrinol Metab*. doi:10.1210/clinem/dqz216

55. Riachi, M., Yilmaz, S., Kurnaz, E., Aycan, Z., Çetinkaya, S., Tranebjærg, L., **Bockenhauer, D** . . . Hussain, K. (2019). Functional Assessment of Variants Associated with Wolfram Syndrome. *Human Molecular Genetics*. doi:10.1093/hmg/ddz212

56. De Rechter, S., **Bockenhauer, D**., Guay-Woodford, L. M., Liu, I., Mallett, A. J., Soliman, N. A., . . . Morawiec-Knysak, A. (2019). ADPedKD: A Global Online Platform on the Management of Children With ADPKD. *Kidney International Reports*, *4*(9), 1271-1284. doi:10.1016/j.ekir.2019.05.015
57. Dufek, S., Cheshire, C., Levine, A. P., Trompeter, R. S., Issler, N., Stubbs, M., . . . **Bockenhauer, D**. (2019). Genetic identification of two novel loci associated with steroid-sensitive nephrotic syndrome. Journal of the American Society of Nephrology : JASN. doi:10.1681/ASN.2018101054

58. Hanson, C. S., Gutman, T., Craig, J. C., Bernays, S., Raman, G., Zhang, Y., **Bockenhauer, D** . . . Tong, A. (2019). Identifying Important Outcomes for Young People With CKD and Their Caregivers: A Nominal Group Technique Study. American Journal of Kidney Diseases. doi:10.1053/j.ajkd.2018.12.040

59. Hayes, W., Longley, C., Scanlon, N., Bryant, W., Stojanovic, J., Kessaris, N., . . . **Bockenhauer, D,** Marks, S. D. (2019). Plasma electrolyte imbalance in pediatric kidney transplant recipients. Pediatric Transplantation. doi:10.1111/petr.13411

60. Adalat, S., Hayes, W. N., Bryant, W. A., Booth, J., Woolf, A. S., Kleta, R., . . . **Bockenhauer, D**. (2019). HNF1B Mutations Are Associated With a Gitelman-like Tubulopathy That Develops During Childhood. Kidney International Reports. doi:10.1016/j.ekir.2019.05.019

61. Wei, W., Tuna, S., Keogh, M. J., Smith, K. R., Aitman, T. J., Beales, P. L., . . . Chinnery, P. F. (2019). Germline selection shapes human mitochondrial DNA diversity.. Science, 364(6442). doi:10.1126/science.aau6520

62. Haffner D, Emma F, Eastwood D, Biosse Duplan M, Bacchetta J, Schnabel D, Wicart P, **Bockenhauer D**, Santos F, Levtchenko E, Harvengt P, Kirchhoff M, Di Rocco F, Chaussain C, Brandi ML, Savendahl L, Briot K, Kamenicky P, Rejnmark L, Linglart A: Clinical practice recommendations for the diagnosis and management of X-linked hypophosphatemia, Nature Reviews Neuroscience, in press

63. Charlotte Gimpel, Carsten Bergmann, **Bockenhauer D**, Luc Breysem, Melissa Cadnapaphornchai, Metin Cetiner, Jan Dudley, F Emma, Martin Konrad, Tess Harris, Peter Harris, Jens König, Max Liebau, Matko Marlais, Djalila Mekahli, Alsion Metcalfe, Jun Oh, Ronald Perrone, Manish Sinha, Andrea Titieni, Roser Torra, Stefanie Weber, Paul Winyard, and Franz Schaefer: International consensus statement on the diagnosis and management of autosomal dominant polycystic kidney disease (ADPKD) in children and young people, Nature Reviews Neuroscience, in press

64. Lopez-Garcia SC, Emma F, Walsh SB, Fila M, Hooman N, Zaniew M, Bertholet-Thomas A, Colussi G, Burgmaier K, Levtchenko E, Sharma J, Singhal J, Soliman NA, Ariceta G, Basu B, Murer L, Tasic V, Tsygin A, Decramer S, Gil-Peña H, Koster-Kamphuis L, La Scola C, Gellermann J, Konrad M, Lilien M, Francisco T, Tramma D, Trnka P, Yüksel S, Caruso MR, Chromek M, Ekinci Z, Gambaro G, Kari JA, König J, Taroni F, Thumfart J, Trepiccione F, Winding L, Wühl E, Ağbaş A, Belkevich A, Vargas-Poussou R, Blanchard A, Conti G, Boyer O, Dursun I, Pınarbaşı AS, Melek E, Miglinas M, Novo R, Mallett A, Milosevic D, Szczepanska M, Wente S, Cheong HI, Sinha R, Gucev Z, Dufek S, Iancu D, Kleta R, Schaefer F, **Bockenhauer D**; European dRTA Consortium: Treatment and long-term outcome in primary distal renal tubular acidosis. Nephrol Dial Transplant. 2019 Feb 18. pii: gfy409. doi: 10.1093/ndt/gfy409. [Epub ahead of print]

65. Dufek S, Holtta T, Trautmann A, Ylinen E, Alpay H, Ariceta G, Aufricht C, Bacchetta J, Bakkaloglu SA, Bayazit A, Cicek RY, Dursun I, Duzova A, Ekim M, Iancu D, Jankauskiene A, Klaus G, Paglialonga F, Pasini A, Printza N, Said Conti V, do Sameiro Faria M, Schmitt CP, Stefanidis CJ, Verrina E, Vidal E, Vondrak K, Webb H, Zampetoglou A, **Bockenhauer D**, Edefonti A, Shroff R: Management of children with congenital nephrotic syndrome: challenging treatment paradigms. Nephrol Dial Transplant. 2018 Jun 21. doi: 10.1093/ndt/gfy165. [Epub ahead of print]

66. Gutman T, Hanson CS, Bernays S, Craig JC, Sinha A, Dart A, Eddy AA, Gipson DS, **Bockenhauer D**, Yap HK, Groothoff J, Zappitelli M, Webb NJA, Alexander SI, Goldstein SL, Furth S, Samuel S, Blydt-Hansen T, Dionne J, Michael M, Wenderfer SE, Winkelmayer WC, Currier H, McTaggart S, Walker A, Ralph AF, Ju A, James LJ, Carter S, Tong A. (2018). Child and parental perspectives on communication and decision-making in pediatric chronic kidney disease: a focus group study. *Am J Kidney Dis. 2018 Oct;72(4):547-559. doi: 10.1053/j.ajkd.2018.05.005. Epub 2018 Jul 3*

67. Biebermann H, Kleinau G, Schnabel D, **Bockenhauer D**, Wilson LC, Tully I, Kiff S, Scheerer P, Reyes M, Paisdzior S, Gregory JW, Allgrove J, Krude H, Mannstadt M, Gardella TJ, Dattani M, Jüppner H, Grüters A: A new multi-system disorder caused by the Gαs mutation p.F376V. *J Clin Endocrinol Metab. 2019 Apr 1;104(4):1079-1089*

68. Schlingmann KP, Bandulik S, Mammen C, Tarailo-Graovac M, Holm R, Baumann M, König J, Lee JJY, Drögemöller B, Imminger K, Beck BB, Altmüller J, Thiele H, Waldegger S, Van't Hoff W, Kleta R, Warth R, van Karnebeek CDM, Vilsen B, **Bockenhauer D**, Konrad M: Germline de-novo mutations in ATP1A1 cause renal hypomagnesemia, refractory seizures and intellectual disability. *Am J Hum Genet. 2018 Nov 1;103(5):808-816*
69. Stiles, C. E., Thursaisingham, R., **Bockenhauer, D**., Platts, L., Kumar, A., & Korbonits, M. (2018). De novo HNF1 homeobox B mutation as a cause for chronic, treatment-resistant hypomagnesaemia. *Endocrinol Diabetes Metab Case Rep. 2018 Mar 21;2018*
70. S Saito H, Noda H, Gatault P, **Bockenhauer D**, Loke KY, Hiort O, Silve C, Sharwood E, Martin RM, Dillon MJ, Gillis D, Harris M, Rao SD, Pauli RM, Gardella TJ, Jüppner H: Progression of Mineral Ion Abnormalities in Patients With Jansen Metaphyseal Chondrodysplasia. *Clin Endocrinol Metab. 2018 Jul 1;103(7):2660-2669*
71. Sharma, S., Ashton, E., Iancu, D., Arthus, M. -F., Hayes, W., Van't Hoff, W., . . . **Bockenhauer, D**. (2018). Long-term outcome in inherited Nephrogenic Diabetes Insipidus. *CKJ: Clinical Kidney Journal*. In press
72. Reichold M, Klootwijk ED, Reinders J, Otto EA, Milani M, Broeker C, Laing C, Wiesner J, Devi S, Zhou W, Schmitt R, Tegtmeier I, Sterner C, Doellerer H, Renner K, Oefner PJ, Dettmer K, Simbuerger JM, Witzgall R, Stanescu HC, Dumitriu S, Iancu D, Patel V, Mozere M, Tekman M, Jaureguiberry G, Issler N, Kesselheim A, Walsh SB, Gale DP, Howie AJ, Martins JR, Hall AM, Kasgharian M, O'Brien K, Ferreira CR, Atwal PS, Jain M, Hammers A, Charles-Edwards G, Choe CU, Isbrandt D, Cebrian-Serrano A, Davies B, Sandford RN, Pugh C, Konecki DS, Povey S, **Bockenhauer D**, Lichter-Konecki U, Gahl WA, Unwin RJ, Warth R, Kleta R: Glycine Amidinotransferase (GATM), Renal Fanconi Syndrome, and Kidney Failure.. *J Am Soc Nephrol. 2018 Jul;29(7):1849-1858*

73. Mozere M, Tekman M, Kari J, **Bockenhauer D**, Kleta R, Stanescu H.: OVAS: an open-source variant analysis suite with inheritance modelling. BMC Bioinformatics. 2018 Feb 8;19(1):46. doi: 10.1186/s12859-018-2030-8

74. Enerbäck S, Nilsson D, Edwards N, Heglind M, Alkanderi S, Ashton E, Deeb A, Kokash FEB, Bakhsh ARA, Van't Hoff W, Walsh SB, D'Arco F, Daryadel A, Bourgeois S, Wagner CA, Kleta R, **Bockenhauer D**, Sayer JA.: Acidosis and Deafness in Patients with Recessive Mutations in FOXI1. J Am Soc Nephrol. 2018 Mar;29(3):1041-1048

75. Walsh PR, Tse Y, Ashton E, Iancu D, Jenkins L, Bienias M, Kleta R, Van't Hoff W, **Bockenhauer D**: Clinical and diagnostic features of Bartter and Gitelman syndromes. Clin Kidney J. 2018 Jun;11(3):302-309

76. Ashton EJ, Legrand A, Benoit V, Roncelin I, Venisse A, Zennaro MC, Jeunemaitre X, Iancu D, Van't Hoff WG, Walsh SB, Godefroid N, Rotthier A, Del Favero J, Devuyst O, Schaefer F, Jenkins LA, Kleta R, Dahan K, Vargas-Poussou R, **Bockenhauer D**.: Simultaneous sequencing of 37 genes identifies causative mutations in the majority of children with renal tubulopathies. Kidney Int. 2018 Feb 1. pii: S0085-2538(17)30796-2. doi: 10.1016/j.kint.2017.10.016. [Epub ahead of print]

77. Warejko JK, Tan W, Daga A, Schapiro D, Lawson JA, Shril S, Lovric S, Ashraf S, Rao J, Hermle T, Jobst-Schwan T, Widmeier E, Majmundar AJ, Schneider R, Gee HY, Schmidt JM, Vivante A, van der Ven AT, Ityel H, Chen J, Sadowski CE, Kohl S, Pabst WL, Nakayama M, Somers MJG, Rodig NM, Daouk G, Baum M, Stein DR, Ferguson MA, Traum AZ, Soliman NA, Kari JA, El Desoky S, Fathy H, Zenker M, Bakkaloglu SA, Müller D, Noyan A, Ozaltin F, Cadnapaphornchai MA, Hashmi S, Hopcian J, Kopp JB, Benador N, **Bockenhauer D**, Bogdanovic R, Stajić N, Chernin G, Ettenger R, Fehrenbach H, Kemper M, Munarriz RL, Podracka L, Büscher R, Serdaroglu E, Tasic V, Mane S, Lifton RP, Braun DA, Hildebrandt F.: Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. Clin J Am Soc Nephrol. 2018 Jan 6;13(1):53-62

78. Aymé S, **Bockenhauer D**, Day S, Devuyst O, Guay-Woodford LM, Ingelfinger JR, Klein JB, Knoers NVAM, Perrone RD, Roberts J, Schaefer F, Torres VE, Cheung M, Wheeler DC, Winkelmayer WC; Conference Participants: Common Elements in Rare Kidney Diseases: Conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney Int. 2017 Oct;92(4):796-808

79. Horn, A., Wright, J., **Bockenhauer, D.**, Van't Hoff, W., & Eastwood, D. M. (2017). The orthopaedic management of lower limb deformity in hypophosphataemic rickets. J Child Orthop. 2017 Aug 1;11(4):298-305.

80. Sawan, Z. A., El-Desoky, S. M., Shalaby, M. A., **Bockenhauer, D.**, & Kari, J. A. (2017). Facial swelling in a child on chronic hemodialysis. Pediatric nephrology (Berlin, Germany), 32(8), 1349-1350

81. Walsh SB, Unwin R, Kleta R, Van't Hoff W, Bass P, Hussain K, Ellard S, **Bockenhauer, D**. (2017). Fainting Fanconi syndrome clarified by proxy: a case report. BMC Nephrol. 2017 Jul 11;18(1):230

82. Chong LSH, Sautenet B, Tong A, Hanson CS, Samuel S, Zappitelli M, Dart A, Furth S, Eddy AA, Groothoff J, Webb NJA, Yap HK, **Bockenhauer** D, Sinha A, Alexander SI, Goldstein SL, Gipson DS, Raman G, Craig JC: Range and Heterogeneity of Outcomes in Randomized Trials of Pediatric Chronic Kidney Disease. J Pediatr. 2017 Apr 24. pii: S0022-3476(17)30383-9

83. Issler N, Dufek S, Kleta R, **Bockenhauer** D, Smeulders N, Van't Hoff W.: Epidemiology of paediatric renal stone disease: a 22-year single centre experience in the UK. BMC Nephrol. 2017 Apr 18;18(1):136

84. Cabezas OR, Flanagan SE, Stanescu H, García-Martínez E, Caswell R, Lango-Allen H, Antón-Gamero M, Argente J, Bussell AM, Brandli A, Cheshire C, Crowne E, Dumitriu S, Drynda R, Hamilton-Shield JP, Hayes W, Hofherr A, Iancu D, Issler N, Jefferies C, Jones P, Johnson M, Kesselheim A, Klootwijk E, Koettgen M, Lewis W, Martos JM, Mozere M, Norman J, Patel V, Parrish A, Pérez-Cerdá C, Pozo J, Rahman SA, Sebire N, Tekman M, Turnpenny PD, Hoff WV, Viering DH, Weedon MN, Wilson P, Guay-Woodford L, Kleta R, Hussain K, Ellard S, **Bockenhauer** D: Polycystic Kidney Disease with Hyperinsulinemic Hypoglycemia Caused by a Promoter Mutation in Phosphomannomutase 2. J Am Soc Nephrol. 2017 Aug;28(8):2529-2539

85. Deschênes G, Vivarelli M, Peruzzi L; ESPN Working Group on Idiopathic Nephrotic Syndrome.: Variability of diagnostic criteria and treatment of idiopathic nephrotic syndrome across European countries. Eur J Pediatr. 2017 Mar 16

86. Besouw MT, Bienias M, Walsh P, Kleta R, Van't Hoff WG, Ashton E, Jenkins L, **Bockenhauer D**.: Clinical and molecular aspects of distal renal tubular acidosis in children. Pediatr Nephrol. 2017 Feb 10. [Epub ahead of print]

87. Sekula P, Li Y, Stanescu HC, Wuttke M, Ekici AB, **Bockenhauer D**, Walz G, Powis SH, Kielstein JT, Brenchley P; GCKD Investigators., Eckardt KU, Kronenberg F, Kleta R, Köttgen A.: Genetic risk variants for membranous nephropathy: extension of and association with other chronic kidney disease aetiologies. Nephrol Dial Transplant. 2017 Feb 1;32(2):325-332

88. Hayes W, Boyle S, Carroll A, **Bockenhauer D**, Marks SD.: Hypomagnesemia and increased risk of new-onset diabetes mellitus after transplantation in pediatric renal transplant recipients. Pediatr Nephrol. 2016 Dec 30 [Epub ahead of print]

89. Blanchard A, **Bockenhauer D**, Bolignano D, Calò LA, Cosyns E, Devuyst O, Ellison DH, Karet Frankl FE, Knoers NV, Konrad M, Lin SH, Vargas-Poussou R.: Gitelman syndrome: consensus and guidance from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney Int. 2017 Jan;91(1):24-33

90. Sawan ZA, El-Desoky SM, Shalaby MA, **Bockenhauer D**, Kari JA.: Facial swelling in a child on chronic hemodialysis. Pediatr Nephrol. 2016 Nov 17 [Epub ahead of print]

91. Dufek S, Booth C, Carroll A, Van't Hoff W, Kleta R, **Bockenhauer D**.: Urea is successful in treating inappropriate antidiuretic hormone secretion in an infant. Acta Paediatr. 2017 Mar;106(3):513-515

92. Caubit X, Gubellini P, Andrieux J, Roubertoux PL, Metwaly M, Jacq B, Fatmi A, Had-Aissouni L, Kwan KY, Salin P, Carlier M, Liedén A, Rudd E, Shinawi M, Vincent-Delorme C, Cuisset JM, Lemaitre MP, Abderrehamane F, Duban B, Lemaitre JF, Woolf AS, **Bockenhauer** D, Severac D, Dubois E, Zhu Y, Sestan N, Garratt AN, Kerkerian-Le Goff L, Fasano L: TSHZ3 deletion causes an autism syndrome and defects in cortical projection neurons. Nat Genet. 2016 Nov;48(11):1359-1369

93. Zaniew M, Bökenkamp A, Kołbuc M, La Scola C, Baronio F, Niemirska A, Szczepańska M, Bürger J, La Manna A, Miklaszewska M, Rogowska-Kalisz A, Gellermann J, Zampetoglou A, Wasilewska A, Roszak M, Moczko J, Krzemień A, Runowski D, Siteń G, Załuska-Leśniewska I, Fonduli P, Zurrida F, Paglialonga F, Gucev Z, Paripovic D, Rus R, Said-Conti V, Sartz L, Chung WY, Park SJ, Lee JW, Park YH, Ahn YH, Sikora P, Stefanidis CJ, Tasic V, Konrad M, Anglani F, Addis M, Cheong HI, Ludwig M, **Bockenhauer D:** Long-term renal outcome in children with OCRL mutations: retrospective analysis of a large international cohort. Nephrol Dial Transplant. 2016 Oct 5. pii: gfw350. [Epub ahead of print]

94. Forsythe E, Sparks K, Best S, Borrows S, Hoskins B, Sabir A, Barrett T, Williams D, Mohammed S, Goldsmith D, Milford DV, **Bockenhauer D**, Foggensteiner L, Beales PL: Risk Factors for Severe Renal Disease in Bardet-Biedl Syndrome. J Am Soc Nephrol. 2016 Sep 22. pii: ASN.2015091029. [Epub ahead of print]

95. Abdelhadi O, Iancu D, Tekman M, Stanescu H, **Bockenhauer D**, Kleta R.: Founder mutation in KCNJ10 in Pakistani patients with EAST syndrome. Mol Genet Genomic Med. 2016 Jun 7;4(5):521-6. doi: 10.1002/mgg3.227

96. Tong A, Samuel S, Zappitelli M, Dart A, Furth S, Eddy A, Groothoff J, Webb NJ, Yap HK, **Bockenhauer D**, Sinha A, Alexander SI, Goldstein SL, Gipson DS, Hanson CS, Evangelidis N, Crowe S, Harris T, Hemmelgarn BR, Manns B, Gill J, Tugwell P, Van Biesen W, Wheeler DC, Winkelmayer WC, Craig JC; SONG-Kids Investigators: Standardised Outcomes in Nephrology-Children and Adolescents (SONG-Kids): a protocol for establishing a core outcome set for children with chronic kidney disease. Trials. 2016 Aug 12;17:401

97. Plumb LA, Van't Hoff W, Kleta R, Reid C, Ashton E, Samuels M, **Bockenhauer D**.: Renal apnoea: extreme disturbance of homoeostasis in a child with Bartter syndrome type IV. Lancet. 2016 Aug 6;388(10044):631-2

98. Sekula P, Li Y, Stanescu HC, Wuttke M, Ekici AB, **Bockenhauer D**, Walz G, Powis SH, Kielstein JT, Brenchley P; GCKD Investigators, Eckardt KU, Kronenberg F, Kleta R, Köttgen A: Genetic risk variants for membranous nephropathy: extension of and association with other chronic kidney disease aetiologies. Nephrol Dial Transplant. 2016 Feb 4. pii: gfw001. [Epub ahead of print]

99. Improda N, Shah P, Güemes M, Gilbert C, Morgan K, Sebire N, **Bockenhauer D**, Hussain K: Hepatocyte Nuclear Factor-4 Alfa Mutation Associated with Hyperinsulinaemic Hypoglycaemia and Atypical Renal Fanconi Syndrome: Expanding the Clinical Phenotype. Horm Res Paediatr. 2016 Jun 1. [Epub ahead of print]

100. Marx-Berger D, Milford DV, Bandhakavi M, Van't Hoff W, Kleta R, Dattani M, **Bockenhauer D:** Tolvaptan is successful in treating inappropriate antidiuretic hormone secretion in infants. Acta Paediatr. 2016 Jul;105(7):e334-7

101. Webb H, Jaureguiberry G, Dufek S, Tullus K, **Bockenhauer D**.: Cyclophosphamide and rituximab in frequently relapsing/steroid-dependent nephrotic syndrome. Pediatr Nephrol. 2016 Apr;31(4):589-94

102. Braun DA, Schueler M, Halbritter J, Gee HY, Porath JD, Lawson JA, Airik R, Shril S, Allen SJ, Stein D, Al Kindy A, Beck BB, Cengiz N, Moorani KN, Ozaltin F, Hashmi S, Sayer JA, **Bockenhauer D**, Soliman NA, Otto EA, Lifton RP, Hildebrandt F. Whole exome sequencing identifies causative mutations in the majority of consanguineous or familial cases with childhood-onset increased renal echogenicity. Kidney Int. 2016 Feb;89(2):468-75

103. Hannan FM, Howles SA, Rogers A, Cranston T, Gorvin CM, Babinsky VN, Reed AA, Thakker CE, **Bockenhauer D**, Brown RS, Connell JM, Cook J, Darzy K, Ehtisham S, Graham U, Hulse T, Hunter SJ, Izatt L, Kumar D, McKenna MJ, McKnight JA, Morrison PJ, Mughal MZ, O'Halloran D, Pearce SH, Porteous ME, Rahman M, Richardson T, Robinson R, Scheers I, Siddique H, Van't Hoff WG, Wang T, Whyte MP, Nesbit MA, Thakker RV.: Adaptor protein-2 sigma subunit mutations causing familial hypocalciuric hypercalcaemia type 3 (FHH3) demonstrate genotype-phenotype correlations, codon bias and dominant-negative effects. Hum Mol Genet. 2015 Sep 15;24(18):5079-92

104. Schueler M, Braun DA, Chandrasekar G, Gee HY, Klasson TD, Halbritter J, Bieder A, Porath JD, Airik R, Zhou W, LoTurco JJ, Che A, Otto EA, **Böckenhauer D**, Sebire NJ, Honzik T, Harris PC, Koon SJ, Gunay-Aygun M, Saunier S, Zerres K, Bruechle NO, Drenth JP, Pelletier L, Tapia-Páez I, Lifton RP, Giles RH, Kere J, Hildebrandt F. DCDC2 mutations cause a renal-hepatic ciliopathy by disrupting Wnt signaling. Am J Hum Genet. 2015 Jan 8;96(1):81-92

85. Recker F, Zaniew M, **Böckenhauer D**, Miglietti N, Bökenkamp A, Moczulska A, Rogowska-Kalisz A, Laube G, Said-Conti V, Kasap-Demir B, Niemirska A, Litwin M, Siteń G, Chrzanowska KH, Krajewska-Walasek M, Sethi SK, Tasic V, Anglani F, Addis M, Wasilewska A, Szczepańska M, Pawlaczyk K, Sikora P, Ludwig M.: Characterization of 28 novel patients expands the mutational and phenotypic spectrum of Lowe syndrome.

Pediatr Nephrol. 2015 Jun;30(6):931-43

86. Sadowski CE, Lovric S, Ashraf S, Pabst WL, Gee HY, Kohl S, Engelmann S, Vega-Warner V, Fang H, Halbritter J, Somers MJ, Tan W, Shril S, Fessi I, Lifton RP, **Bockenhauer D**, El-Desoky S, Kari JA, Zenker M, Kemper MJ, Mueller D, Fathy HM, Soliman NA; the SRNS Study Group, Hildebrandt F.: A Single-Gene Cause in 29.5% of Cases of Steroid-Resistant Nephrotic Syndrome. J Am Soc Nephrol. 2015 Jun;26(6):1279-89

107. Simpkin A, Cochran E, Cameron F, Dattani M, de Bock M, Dunger DB, Forsander G, Guran T, Harris J, Isaac I, Hussain K, Kleta R, Peters C, Tasic V, Williams R, Yap Kok Peng F, O'Rahilly S, Gorden P, Semple RK, **Bockenhauer D**.: Insulin Receptor and the Kidney: Nephrocalcinosis in Patients with Recessive INSR Mutations. Nephron Physiol. 2014 Oct 24. [Epub ahead of print]

108. Webb NJ, Frew E, Brettell EA, Milford DV, **Bockenhauer D**, Saleem MA, Christian M, Hall AS, Koziell A, Maxwell H, Hegde S, Finlay ER, Gilbert RD, Booth J, Jones C, McKeever K, Cook W, Ives NJ; PREDNOS 2 study group.: Short course daily prednisolone therapy during an upper respiratory tract infection in children with relapsing steroid-sensitive nephrotic syndrome (PREDNOS 2): protocol for a randomised controlled trial. Trials. 2014 Apr 27;15:147

109. Kari JA, El Desoky SM, Singh AK, Gari MA, Kleta R, **Bockenhauer D**.: The case | Renal tubular acidosis and eye findings. Kidney Int. 2014 Jul;86(1):217-8

110. Kari JA, Montini G, **Bockenhauer D,** Brennan E, Rees L, Trompeter RS, Tullus K, Van't Hoff W, Waters A, Ashton E, Lench N, Sebire NJ, Marks SD.: Clinico-pathological correlations of congenital and infantile nephrotic syndrome over twenty years. Pediatr Nephrol. 2014 Nov;29(11):2173-80

111. Safder O, El-Desoky SM, **Bockenhauer D**, Sabire N, Kari JA: Steroid-resistant nephrotic syndrome in a child with dysmorphic features. Pediatr Nephrol. 2014 May;29(5):839-40

112. Kari JA, El-Desoky SM, Gari M, Malik K, Vega-Warner V, Lovric S, **Bockenhauer D**: Steroid-resistant nephrotic syndrome: impact of genetic testing. Ann Saudi Med. 2013 Nov-Dec;33(6):533-8

113. Klootwijk ED, Reichold M, Helip-Wooley A, Tolaymat A, Broeker C, Robinette SL, Reinders J, Peindl D, Renner K, Eberhart K, Assmann N, Oefner PJ, Dettmer K, Sterner C, Schroeder J, Zorger N, Witzgall R, Reinhold SW, Stanescu HC, **Bockenhauer D**, Jaureguiberry G, Courtneidge H, Hall AM, Wijeyesekera AD, Holmes E, Nicholson JK, O'Brien K, Bernardini I, Krasnewich DM, Arcos-Burgos M, Izumi Y, Nonoguchi H, Jia Y, Reddy JK, Ilyas M, Unwin RJ, Gahl WA, Warth R, Kleta R.: Mistargeting of peroxisomal EHHADH and inherited renal Fanconi's syndrome. N Engl J Med. 2014 Jan 9;370(2):129-38

114. Ashraf S, Gee HY, Woerner S, Xie LX, Vega-Warner V, Lovric S, Fang H, Song X, Cattran DC, Avila-Casado C, Paterson AD, Nitschké P, Bole-Feysot C, Cochat P, Esteve-Rudd J, Haberberger B, Allen SJ, Zhou W, Airik R, Otto EA, Barua M, Al-Hamed MH, Kari JA, Evans J, Bierzynska A, Saleem MA, **Böckenhauer D**, Kleta R, El Desoky S, Hacihamdioglu DO, Gok F, Washburn J, Wiggins RC, Choi M, Lifton RP, Levy S, Han Z, Salviati L, Prokisch H, Williams DS, Pollak M, Clarke CF, Pei Y, Antignac C, Hildebrandt F.: ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. J Clin Invest. 2013 Dec 2;123(12):5179-89

115. Kari JA, **Bockenhauer D**, Stanescu H, Gari M, Kleta R, Singh AK: Consanguinity in Saudi Arabia: A Unique Opportunity for Pediatric Kidney Research. Am J Kidney Dis. 2013 Nov 13

116. Zdebik AA, Mahmood F, Stanescu HC, Kleta R, **Bockenhauer D**, Russell C.: Epilepsy in kcnj10 Morphant Zebrafish Assessed with a Novel Method for Long-Term EEG Recordings. PLoS One. 2013 Nov 14;8(11):e79765

117. Parrock S, Hussain S, Issler N, Differ AM, Lench N, Guarino S, Oosterveld MJ, Keijzer-Veen M, Brilstra E, van Wieringen H, Konijnenberg AY, Amin-Rasip S, Dumitriu S, Klootwijk E, Knoers N, **Bockenhauer D**, Kleta R, Zdebik AA: KCNJ10 Mutations Display Differential Sensitivity to Heteromerisation with KCNJ16.Nephron Physiol. 2013 Nov 2;123(3-4):7-14

118. Bausch B, Wellner U, Bausch D, Schiavi F, Barontini M, Sanso G, Walz MK, Peczkowska M, Weryha G, Dall'igna P, Cecchetto G, Bisogno G, Moeller L, **Bockenhauer D,** Patocs A, Racz K, Zabolotnyi D, Yaremchuk S, Dzivite-Krisane I, Castinetti F, Taieb D, Malinoc A, von Dobschuetz E, Roessler J, Schmid KW, Opocher G, Eng C, Neumann HP.: Long term prognosis of patients with pediatric pheochromocytoma. Endocr Relat Cancer. 2013 Dec 16;21(1):17-25

119. Schmidts M, Vodopiutz J, Christou-Savina S, Cortés CR, McInerney-Leo AM, Emes RD, Arts HH, Tüysüz B, D'Silva J, Leo PJ, Giles TC, Oud MM, Harris JA, Koopmans M, Marshall M, Elçioglu N, Kuechler A, **Bockenhauer D**, Moore AT, Wilson LC, Janecke AR, Hurles ME, Emmet W, Gardiner B, Streubel B, Dopita B, Zankl A, Kayserili H, Scambler PJ, Brown MA, Beales PL, Wicking C; UK10K, Duncan EL, Mitchison HM: Mutations in the Gene Encoding IFT Dynein Complex Component WDR34 Cause Jeune Asphyxiating Thoracic Dystrophy.Am J Hum Genet. 2013 Nov 7;93(5):932-44.

120. El-Desoky SM, **Bockenhauer D**, Shroff R, Kari JA.: Subcutaneous nodules in a child on long-term dialysis:

Answers. Pediatr Nephrol. 2013 Aug 30. [Epub ahead of print]

121. Cross JH, Arora R, Heckemann RA, Gunny R, Chong K, Carr L, Baldeweg T, Differ AM, Lench N, Varadkar S, Sirimanna T, Wassmer E, Hulton SA, Ognjanovic M, Ramesh V, Feather S, Kleta R, Hammers A, **Bockenhauer D**: Neurological features of epilepsy, ataxia, sensorineural deafness, tubulopathy syndrome. Dev Med Child Neurol. 2013 Sep;55(9):846-56.

122. Hoff S, Halbritter J, Epting D, Frank V, Nguyen TM, van Reeuwijk J, Boehlke C, Schell C, Yasunaga T, Helmstädter M, Mergen M, Filhol E, Boldt K, Horn N, Ueffing M, Otto EA, Eisenberger T, Elting MW, van Wijk JA, **Bockenhauer D**, Sebire NJ, Rittig S, Vyberg M, Ring T, Pohl M, Pape L, Neuhaus TJ, Elshakhs NA, Koon SJ, Harris PC, Grahammer F, Huber TB, Kuehn EW, Kramer-Zucker A, Bolz HJ, Roepman R, Saunier S, Walz G, Hildebrandt F, Bergmann C, Lienkamp SS. ANKS6 is a central component of a nephronophthisis module linking NEK8 to INVS and NPHP3. Nat Genet. 2013 Aug;45(8):951-6

123. Shute, R., Jeelani, O., Lee, L., Brennan, E., **Bockenhauer, D**., Barnicoat, A., Shroff, R. (2013). Rapid head growth in a baby with autosomal dominant polycystic kidney disease (ADPKD) Pediatr Nephrol. 2014 Feb;29(2):219-21

124. Coenen MJ, Hofstra JM, Debiec H, Stanescu HC, Medlar AJ, Stengel B, Boland-Augé A, Groothuismink JM, **Bockenhauer D**, Powis SH, Mathieson PW, Brenchley PE, Kleta R, Wetzels JF, Ronco P: Phospholipase A2 receptor (PLA2R1) sequence variations in idiopathic membranous nephropathy; J Am Soc Nephrol. 2013 Mar;24(4):677-83

125. Mahmood F, Mozeres M, Zdebik AA, Stanescu HC, Tobin J, Beales PL, Kleta R, Bockenhauer D, Russell C: Generation and validation of a zebrafish model of EAST (epilepsy, ataxia, sensorineural deafness and tubulopathy) syndrome, Dis Mod Mech *6*(3), 652-660.

126. Pasternack SM, **Böckenhauer D,** Refke M, Tasic V, Draaken M, Conrad C, Born M, Betz RC, Reutter H, Ludwig M.: A Premature Termination Mutation in a Patient with Lowe Syndrome without Congenital Cataracts: Dropping the "O" in OCRL, Klin Padiatr. 2013 Jan;225(1):29-33.

127. G Jaureguiberry, D Parry, M Quentric, N Himmerkus, T Koike, J Poulter, E Klootwijk, SL Robinette, AJ Howie, V Patel, M-L Figueres, HC Stanescu, N Issler, JK Nicholson, **D Bockenhauer**, C Laing, SB Walsh, DA McCredie, S Povey, A Asselin, A Picard, A Coulomb, AJ Medlar, I Bailleul-Forestier, A Verloes, C Le Caignec, G Roussey, J Guiol, B Isidor, C Logan, R Shore, C Johnson, C Inglehearn, S Al-Bahlani, M Schmittbuhl, F Clauss, M Huckert, V Laugel, E Ginglinger, S Pajarola, G Spartà, D Bartholdi, A Rauch, M-C Addor, PM Yamaguti, HP Safatle, AC Acevedo, H Martelli-Júnior, PE dos Santos Netos, RD Coletta, S Gruessel, C Sandmann, D Ruehmann, CB Langman, SJ Scheinman, D Ozdemir-Ozenen, TC Hart, PS Hart, U Neugebauer, E Schlatter, P Houillier, WA Gahl, M Vikkula, A Bloch-Zupan, M Bleich, H Kitagawa, RJ Unwin, A Mighell, A Berdal, R Kleta: Nephrocalcinosis (Enamel Renal Syndrome) Caused by Autosomal Recessive FAM20A Mutations; Nephron Physiology 2012;122 (1-2), 1-6

128. DG Bichet, A El Tarazi, J Matar, Y Lussier, MF Arthus, M Lonergan, **D Bockenhauer** and P Bissonnette: Aquaporin-2: new mutations responsible for autosomal-recessive nephrogenic diabetes insipidus—update and epidemiology; Clinical Kidney Journal 2012;5 (3), 195-202

129. Ovunc B, Ashraf S, Vega-Warner V, **Bockenhauer D**, Soliman Elshakhs NA, Joseph M, Hildebrandt F: Mutation Analysis of NPHS1 in a Worldwide Cohort of Congenital Nephrotic Syndrome Patients, Nephron Clin Pract 2012;**120**(3):c139-c146

130. Has C, Spartà G, Kirits D, Weibel L, Moeller A, Vega-Warner V, Waters A, He Y, Anikster Y, Esser P, Straub B, Hausser I, **Bockenhauer D**, Dekel B, Hildebrandt F, Bruckner-Tuderman L, and Laube GF: Integrin □3 Mutations with Kidney, Lung and Skin Disease, New Engl J Med 2012, Apr 19;366(16):1508-14

131. Williams EL, **Bockenhauer D**, Van't Hoff WG, Johri N, Laing C, Sinha MD, Unwin R, Viljoen A, Rumsby G: The enzyme 4-hydroxy-2-oxoglutarate aldolase is deficient in primary hyperoxaluria type 3; Nephrol Dial Transplant 2012 Aug;27(8):3191-5

132. Boyden LM, Choi M, Choate KA, Nelson-Williams CJ, Farhi A, Toka HR, Tikhonova IR, Bjornson R, Mane SM, Colussi G, Lebel M, Gordon RD, Semmekrot BA, Poujol A, Välimäki MJ, De Ferrari ME, Sanjad SA, Gutkin M, Karet FE, Tucci JR, Stockigt JR, Keppler-Noreuil KM, Porter CC, Anand SK, Whiteford ML, Davis ID, Dewar SB, Bettinelli A, Fadrowski JJ, Belsha CW, Hunley TE, Nelson RD, Trachtman H, Cole TR, Pinsk M, **Bockenhauer D**, Shenoy M, Vaidyanathan P, Foreman JW, Rasoulpour M, Thameem F, Al-Shahrouri HZ, Radhakrishnan J, Gharavi AG, Goilav B, Lifton RP: Mutations in kelch-like 3 and cullin 3 cause hypertension and electrolyte abnormalities. Nature. 2012 Jan 22;482(7383):98-102

133. **Bockenhauer D**, Penney MD, Hampton D, van't Hoff W, Gullett A, Sailesh S, Bichet DG: A Family With Hyponatremia and the Nephrogenic Syndrome of Inappropriate Antidiuresis, American Journal of Kidney Diseases, 2012 Apr;59(4):566-8

134. Quinlan C, Guegan K, Offiah A, O"Neill RJ, Hiorns MP, Ellard S, **Bockenhauer D**, van't Hoff W and Waters A: Growth in PHEX-associated X-linked hypophosphataemic rickets: the importance of early treatment. Ped Neph, 2012 Apr;27(4):581-8

135. Vivante, A., Lotan, D., Pode-Shakked, N., Landau, D., Svec, P., Nampoothiri, S., Verma, I., Abu-Libdeh,

A., **Bockenhauer, D**., Dekel, B., Anikster, Y. (2011). Familial Autosomal Recessive Renal Tubular Acidosis: Importance of Early Diagnosis. *Nephron Physiol* 119(3), p31-p39

136. Barber, B. R., Weber, M. A., **Bockenhauer, D**., Hiorns, M. P., McHugh, K. (2011). Postmortem MRI of bladder agenesis. *Pediatr Radiol* 41(1), 110-112

137. Freudenthal, B., Kulaveerasingam, D., Lingappa, L., Shah, M. A., Brueton, L., Wassmer, E., Ognjanovic, M., Dorison, N., Reichold, M., **Bockenhauer, D**., Kleta, R., Zdebik, A. A. (2011). KCNJ10 Mutations Disrupt Function in Patients with EAST Syndrome. *Nephron Physiol* 119(3), p40-p48

138. Jaureguiberry, G., Van't Hoff, W., Mushtaq, I., Desai, D., Mann, N. P., Kleta, R., Bichet, D. G., **Bockenhauer, D**. (2011). A patient with polyuria and hydronephrosis. *Pediatr Nephrol* 26(11), 1977-1980

139. Kasperavičiūtė, D., Catarino, C. B., Chinthapalli, K., Clayton, L. M., Thom, M., Martinian, L., Cohen, H., Adalat, S., **Bockenhauer, D.**, Pope, S. A., Lench, N., Koltzenburg, M., Duncan, J. S., Hammond, P., Hennekam, R. C., Land, J. M., Sisodiya, S. M. (2011). Uncovering genomic causes of co-morbidity in epilepsy: gene-driven phenotypic characterization of rare microdeletions. *PLoS One* 6(8), e23182-

140. Kenny, J., Lees, M. M., Drury, S., Barnicoat, A., Van't Hoff, W., Palmer, R., Morrogh, D., Waters, J. J., Lench, N. J., **Bockenhauer, D.** (2011). Sotos syndrome, infantile hypercalcemia, and nephrocalcinosis: a contiguous gene syndrome. *Pediatr Nephrol* 26(8), 1331-1334

141. Otto, E. A., Ramaswami, G., Janssen, S., Chaki, M., Allen, S. J., Zhou, W., Airik, R., Hurd, T. W., Ghosh, A. K., Wolf, M. T., Hoppe, B., Neuhaus, T. J., **Bockenhauer, D.,** Milford, D. V., Soliman, N. A., Antignac, C., Saunier, S., Johnson, C. A., Hildebrandt, F., GPN Study Group, (2011). Mutation analysis of 18 nephronophthisis associated ciliopathy disease genes using a DNA pooling and next generation sequencing strategy. *J Med Genet* 48(2), 105-116

142. Stanescu, H. C., Arcos-Burgos, M., Medlar, A., **Bockenhauer, D.**, Kottgen, A., Dragomirescu, L., Voinescu, C., Patel, N., Pearce, K., Hubank, M., Stephens, H. A., Laundy, V., Padmanabhan, S., Zawadzka, A., Hofstra, J. M., Coenen, M. J., den Heijer, M., Kiemeney, L. A., Bacq-Daian, D., Stengel, B., Powis, S. H., Brenchley, P., Feehally, J., Rees, A. J., Debiec, H., Wetzels, J. F., Ronco, P., Mathieson, P. W., Kleta, R. (2011). Risk HLA-DQA1 and PLA(2)R1 alleles in idiopathic membranous nephropathy. *New England Journal of Medicine* 364(7), 616-26

143. Thompson, D. A., Feather, S., Stanescu, H. C., Freudenthal, B., Zdebik, A. A., Warth, R., Ognjanovic, M., Hulton, S. A., Wassmer, E., van't Hoff, W., Russell-Eggitt, I., Dobbie, A., Sheridan, E., Kleta, R., **Bockenhauer D**. (2011). Altered electroretinograms in patients with KCNJ10 mutations and EAST syndrome. *J Physiol* 589(Pt 7), 1681-1689

144. **Bockenhauer D**, Bökenkamp A, Nuutinen M, Unwin R, van't Hoff W, Sirimanna T, Vrljicak K, Ludwig M: Novel *OCRL* mutations in patients with Dent-2 disease, J Ped Gen, 2012 1(1) 15-23

145. **Bockenhauer, D.**, van't Hoff, W., Dattani, M., Lehnhardt, A., Subtirelu, M., Hildebrandt, F., Bichet, D. G. (2010). Secondary nephrogenic diabetes insipidus as a complication of inherited renal diseases. *Nephron Physiol* 116(4), p23-p29

146. Matejas, V., Hinkes, B., Alkandari, F., Al-Gazali, L., Annexstad, E., Aytac, M. B., Barrow, M., Bláhová, K., **Bockenhauer, D**., Cheong, H. I., Maruniak-Chudek, I., Cochat, P., Dötsch, J., Gajjar, P., Hennekam, R. C., Janssen, F., Kagan, M., Kariminejad, A., Kemper, M. J., Koenig, J., Kogan, J., Kroes, H. Y., Kuwertz-Bröking, E., Lewanda, A. F., Medeira, A., Muscheites, J., Niaudet, P., Pierson, M., Saggar, A., Seaver, L., Suri, M., Tsygin, A., Wühl, E., Zurowska, A., Uebe, S., Hildebrandt, F., Antignac, C., Zenker, M. (2010). Mutations in the human laminin beta2 (LAMB2) gene and the associated phenotypic spectrum. *Hum Mutat* 31(9), 992-1002

147. Mekahli, D., Woolf, A. S., **Bockenhauer, D.** (2010). Similar renal outcomes in children with ADPKD diagnosed by screening or presenting with symptoms. *Pediatr Nephrol* 25(11), 2275-2282

148. Reichold, M., Zdebik, A. A., Lieberer, E., Rapedius, M., Schmidt, K., Bandulik, S., Sterner, C., Tegtmeier, I., Penton, D., Baukrowitz, T., Hulton, S. A., Witzgall, R., Ben-Zeev, B., Howie, A. J., Kleta, R., Bockenhauer, D., Warth, R. (2010). KCNJ10 gene mutations causing EAST syndrome (epilepsy, ataxia, sensorineural deafness, and tubulopathy) disrupt channel function. *Proc Natl Acad Sci U S A* 107(32), 14490-14495

149. Schoeb, D. S., Chernin, G., Heeringa, S. F., Matejas, V., Held, S., Vega-Warner, V., **Bockenhauer, D.**, Vlangos, C. N., Moorani, K. N., Neuhaus, T. J., Kari, J. A., MacDonald, J., Saisawat, P., Ashraf, S., Ovunc, B., Zenker, M., Hildebrandt, F., Gesellschaft für Paediatrische Nephrologie (GPN) Study Group, (2010). Nineteen novel NPHS1 mutations in a worldwide cohort of patients with congenital nephrotic syndrome (CNS). *Nephrol Dial Transplant* 25(9), 2970-2976

150. Waters, A. M., Pappworth, I., Marchbank, K., **Bockenhauer, D.**, Tullus, K., Pickering, M. C., Strain, L., Sebire, N., Shroff, R., Marks, S. D., Goodship, T. H., Rees, L. (2010). Successful renal transplantation in factor H autoantibody associated HUS with CFHR1 and 3 deficiency and CFH variant G2850T. *Am J Transplant* 10(1), 168-172

151. Adalat, S., Woolf, A. S., Johnstone, K. A., Wirsing, A., Harries, L. W., Long, D. A., Hennekam, R. C., Ledermann, S. E., Rees, L., van't Hoff, W., Marks, S. D., Trompeter, R. S., Tullus, K., Winyard, P. J., Cansick, J., Mushtaq, I., Dhillon, H. K., Bingham, C., Edghill, E. L., Shroff, R., Stanescu, H., Ryffel, G. U., Ellard, S., **Bockenhauer, D.** (2009). HNF1B mutations associate with hypomagnesemia and renal magnesium wasting. *J Am Soc Nephrol* 20(5), 1123-1131

152. **Bockenhauer, D.**, Carpentier, E., Rochdi, D., Van't Hoff, W., Breton, B., Bernier, V., Bouvier, M., Bichet, D. G. (2009). Vasopressin Type 2 Receptor V88M Mutation: Molecular Basis of Partial and Complete Nephrogenic Diabetes Insipidus. *Nephron Physiol* 114(1), p1-p10

153. **Bockenhauer, D.**, Feather, S., Stanescu, H. C., Bandulik, S., Zdebik, A. A., Reichold, M., Tobin, J., Lieberer, E., Sterner, C., Landoure, G., Ruchi, A., Sirimanna, T., Thompson, D., Cross, J. H., van't Hoff, W., Al Masri, O., Tullus, K., Yeung, S., Anikster, Y., Klootwijk, E., Hubank, M., Dillon, M. J., Heitzmann, D., Arcos-Burgos, M., Knepper, M. A., Dobbie, A., Gahl, W. A., Warth, R., Sheridan, E., Kleta, R. (2009). Epilepsy, ataxia, sensorineural deafness, tubulopathy, and KCNJ10 mutations. *New England Journal of Medicine* 360(19), 1960-197

154. **Bockenhauer, D.**, van't Hoff, W., Chernin, G., Heeringa, S. F., Sebire, N. J. (2009). Membranoproliferative glomerulonephritis associated with a mutation in Wilms' tumour suppressor gene 1. *Pediatr Nephrol* 24(7), 1399-1401

155. Bokenkamp, A., **Bockenhauer, D.,** Cheong, H. I., Hoppe, B., Tasic, V., Unwin, R., Ludwig, M. (2009). Dent-2 Disease: A Mild Variant of Lowe Syndrome. *Journal of Pediatrics* 155(1), 94-99

156. Cansick, J., Rees, L., Koffman, G., Van't Hoff, W., Bockenhauer, D. (2009). A fatal case of cerebral oedema with hyponatraemia and massive polyuria after renal transplantation. *Pediatr Nephrol* 2009 Jun;24(6):1231-4

157. Krischock, L., Gullett, A., **Bockenhauer, D.**, Rees, L., Trompeter, R. S., Marks, S. D. (2009). Calcineurin-inhibitor free immunosuppression with mycophenolate mofetil and corticosteroids in paediatric renal transplantation improves renal allograft function without increasing acute rejection. *Pediatr Transplant* 13(4), 475-481

158. **Bockenhauer, D.**, Bokenkamp, A., van't Hoff, W., Levtchenko, E., Kist-van Holthe, J. E., Tasic, V., Ludwig, M. (2008). Renal phenotype in Lowe Syndrome: a selective proximal tubular dysfunction. *Clin J Am Soc Nephrol* 3(5), 1430-1436

159. **Bockenhauer, D.,** Cruwys, M., Kleta, R., Halperin, L. F., Wildgoose, P., Souma, T., Nukiwa, N., Cheema-Dhadli, S., Chong, C. K., Kamel, K. S., Davids, M. R., Halperin, M. L. (2008). Antenatal Bartter's syndrome: why is this not a lethal condition? *QJM* 101, 927-942

160. **Bockenhauer, D**., Debiec, H., Sebire, N., Barratt, M., Warwicker, P., Ronco, P., Kleta, R. (2008). Familial membranous nephropathy: an X-linked genetic susceptibility? *Nephron Clinical Practice* 108(1), c10-c15

161. **Bockenhauer, D.**, Rees, L., Neumann, H., Foo, Y. (2008). A sporadic case of paraganglioma undetected by urine metabolite screening. *Pediatr Nephrol* 2008 Oct;23(10):1889-91

162. Bredrup, C., Matejas, V., Barrow, M., Blahova, K., **Bockenhauer, D**., Fowler, D. J., Gregson, R. M., Maruniak-Chudek, I., Medeira, A., Mendonca, E. L., Kagan, M., Koenig, J., Krastel, H., Kroes, H. Y., Saggar, A., Sawyer, T., Schittkowski, M., Swietlinski, J., Thompson, D., Vandevoorde, R. G., Wittebol-Post, D., Woodruff, G., Zurowska, A., Hennekam, R. C., Zenker, M., Russell-Eggitt, I. (2008). Ophthalmological Aspects of Pierson Syndrome. *Am J Ophthalmol.*

163. **Bockenhauer, D.**, Rees, L., van't, H. W. (2006). An association of tubular dysfunction, cortical macrocysts and chronic kidney disease. *Pediatr Nephrol* 21(4), 580-583

164. Muller, D., Kausalya, P. J., **Bockenhauer, D.,** Thumfart, J., Meij, I. C., Dillon, M. J., van't, H. W., Hunziker, W. (2006). Unusual clinical presentation and possible rescue of a novel claudin-16 mutation. *Journal of Clinical Endocrinology and Metabolism* 91(8), 3076-3079

165. Sebire, N. J., **Bockenhauer, D.** (2005). Posttransplant de novo membranous nephropathy in childhood. *Fetal and Pediatric Pathology* 24(2), 95-103

166. Sreedharan, R., **Bockenhauer, D.** (2005). Congenital nephrotic syndrome responsive to angiotensin-converting enzyme inhibition. *Pediatr Nephrol* 20(9), 1340-1342

167. Kleta, R., Romeo, E., Ristic, Z., Ohura, T., Stuart, C., rcos-Burgos, M., Dave, M. H., Wagner, C. A., Camargo, S. R., Inoue, S., Matsuura, N., Helip-Wooley, A., **Bockenhauer, D**., Warth, R., Bernardini, I., Visser, G., Eggermann, T., Lee, P., Chairoungdua, A., Jutabha, P., Babu, E., Nilwarangkoon, S., Anzai, N., Kanai, Y., Verrey, F., Gahl, W. A., Koizumi, A. (2004). Mutations in SLC6A19, encoding B0AT1, cause Hartnup disorder. *Nature Genetics* 36(9), 999-1002

168. Lechner, B. L., **Bockenhauer, D**., Iragorri, S., Kennedy, T. L., Siegel, N. J. (2004). The risk of cardiovascular disease in adults who have had childhood nephrotic syndrome. *Pediatr Nephrol* 19(7), 744-748

169. Levy, D. I., Velazquez, H., Goldstein, S. A., **Bockenhauer, D**. (2004). Segment-specific expression of 2P domain potassium channel genes in human nephron. *Kidney Int* 65(3), 918-926

170. **Bockenhauer, D**., Zilberberg, N., Goldstein, S. A. (2001). KCNK2: reversible conversion of a hippocampal potassium leak into a voltage-dependent channel. *Nature Neuroscience* 4(5), 457-458

171. Hausladen, J., Granahan, E., **Bockenhauer, D**. (2001). Case report: Teenage girl with proteinuria and amenorrhea. *Curr Opin Pediatr* 13(2), 150-153

172. **Bockenhauer, D.**, Nimmakayalu, M. A., Ward, D. C., Goldstein, S. A., Gallagher, P. G. (2000). Genomic organization and chromosomal localization of the murine 2 P domain potassium channel gene Kcnk8: conservation of gene structure in 2 P domain potassium channels. *Gene* 261(2), 365-372

173. Jorquera, P., Wu, J., **Bockenhauer, D.** (1999). Nonanion gap metabolic acidosis in a newborn. *Curr Opin Pediatr* 11(2), 169-173

Other publications

87. Tong A, Manns B, Wang AYM, Hemmelgarn B, Wheeler DC, Gill J, Tugwell P, Pecoits-Filho R, Crowe S, Harris T, Van Biesen W, Winkelmayer WC, Levin A, Thompson A, Perkovic V, Ju A, Gutman T, Bernier-Jean A, Viecelli AK, O'Lone E, Shen J, Josephson MA, Cho Y, Johnson DW, Sautenet B, Tonelli M, Craig JC; SONG Implementation Workshop Investigators: Implementing core outcomes in kidney disease: report of the Standardized Outcomes in Nephrology (SONG) implementation workshop. *Kidney Int. 2018 Dec;94(6):1053-1068*

88. Lambe T, Frew E, Ives NJ, Woolley RL, Cummins C, Brettell EA, Barsoum EN, Webb NJA; PREDNOS Trial Team.: Mapping the Paediatric Quality of Life Inventory (PedsQL™) Generic Core Scales onto the Child Health Utility Index-9 Dimension (CHU-9D) Score for Economic Evaluation in Children. Pharmacoeconomics. 2017 Dec 20. doi: 10.1007/s40273-017-0600-7. [Epub ahead of print]

89. Deschênes et al: Variability of diagnostic criteria and treatment of idiopathic nephrotic syndrome across European countries. Eur J Pediatr. 2017 May;176(5):647-654

177. Lethaby D, **Bockenhauer D**, Cyriac J: Is the use of Furosemide beneficial in the treatment of acute Kidney injury in the paediatric population including neonates? Arch Dis Child. 2015 Jul;100(7):713-5. doi: 10.1136/archdischild-2015-308472

178. Guay-Woodford LM, Bissler JJ, Braun MC, **Bockenhauer D**, Cadnapaphornchai MA, Dell KM, Kerecuk L, Liebau MC, Alonso-Peclet MH, Shneider B, Emre S, Heller T, Kamath BM, Murray KF, Moise K, Eichenwald EE, Evans J, Keller RL, Wilkins-Haug L, Bergmann C, Gunay-Aygun M, Hooper SR, Hardy KK, Hartung EA, Streisand R, Perrone R, Moxey-Mims M.: Consensus expert recommendations for the diagnosis and management of autosomal recessive polycystic kidney disease: report of an international conference. J Pediatr. 2014 Sep;165(3):611-7

179. Heeringa et al: Thirteen novel NPHS1 mutations in a large cohort of children with congenital nephrotic syndrome. Nephrol Dial Transplant. 2008 Nov;23(11):3527-33. (Paper resulting from participation in a multicentre study on congenital nephrotic syndrome)

180. **Bockenhauer, D**., Stanescu, H. C., Kleta, R. (2009). The EAST syndrome and KCNJ10 mutations. New England Journal of Medicine 361, 630-631 (Reply to a letter to the editor)

181. Erlic et al: Clinical predictors and algorithm for the genetic diagnosis of pheochromocytoma patients. Clin Cancer Res 2009 Oct 15; 15(20):6378-85 (Paper resulting from participation in a multicentre study on pheochromocytoma)

182. Chernin G, Vega-Warner V, Schoeb DS, Heeringa SF, Ovunc B, Saisawat P, Cleper R, Ozaltin F, Hildebrandt F; Members of the GPN Study Group Genotype/phenotype correlation in nephrotic syndrome caused by WT1 mutations. Clin J Am Soc Nephrol. 2010 Sep;5(9):1655-62 (Paper resulting from participation in a multicentre study on nephrotic syndrome)

183. **Bockenhauer D**: Pulse wave velocity, blood pressure and bicycle tyres Ped Neph, 2011; 26(8); 1343 (letter to the editor)

Reviews and editorials

1. Bockenhauer, D., & Florio, G. (2024). Ascites: Under- and Overfill Is Amiloride the Answer?. *JOURNAL OF THE AMERICAN SOCIETY OF NEPHROLOGY*, *35*(11), 1453-1455. doi:10.1681/ASN.0000000000000493
2. Bockenhauer, D., & Stanescu, H. (2024). Distal renal tubular acidosis and WDR72: some answers, more

questions.. *Pediatric Nephrology*. doi:10.1007/s00467-024-06504-4
3. Bichet, D. G., & Bockenhauer, D. (2024). Thirst, Hunger, and Nephrogenic Diabetes Insipidus.. *New England Journal of Medicine*, *390*(20), 1922-1924. doi:10.1056/NEJMcibr2400066
4. Bockenhauer, D., & Boyer, O. (2024). Less or later: Treatment dilemmas in congenital nephrotic syndrome. *Acta Paediatrica: Nurturing the Child*, 2 pages. doi:10.1111/apa.17310
5. **Bockenhauer, D.**, Knoers, N. V. A. M., & Bichet, D. G. (2022). What's in a name? That which we call diabetes does not taste sweet!. *Pediatric Nephrology*. doi:10.1007/s00467-022-05815-8
6. Walker, E., Hayes, W., & Bockenhauer, D. (2024). Inherited non-FGF23-mediated phosphaturic disorders: A kidney-centric review. *BEST PRACTICE & RESEARCH CLINICAL ENDOCRINOLOGY & METABOLISM*, *38*(2), 12 pages. doi:10.1016/j.beem.2023.101843
7. de Baaij, J. H. F., **Bockenhauer, D.**, Claverie-Martin, F., Hoenderop, J. G. J., Hoorn, E. J., Houillier, P., . . . Vargas Poussou, R. (2022). Comment to "Recommendation on an updated standardization of serum magnesium reference ranges".. *European Journal of Nutrition*. doi:10.1007/s00394-022-03004-9
8. Bockenhauer D, Kleta R. Tubulopathy meets Sherlock Holmes: biochemical fingerprinting of disorders of altered kidney tubular salt handling PEDIATRIC NEPHROLOGY 36(8):2553-2561 18 Jun 2021
9. Dufek-Kamperis, S., Kleta, R., **Bockenhauer, D.**, Gale, D., & Downie, M. L. (2020). Novel insights in the genetics of steroid-sensitive nephrotic syndrome in childhood. Pediatr Nephrol. doi:10.1007/s00467-020-04780-4
10. Downie, M. L., Lopez Garcia, S. C., Kleta, R., & **Bockenhauer, D**. (2020). Inherited Tubulopathies of the Kidney: Insights from Genetics. Clin J Am Soc Nephrol. doi:10.2215/CJN.14481119
11. Gale, D., Mallett, A., Patel, C., Sneddon, T. P., Rehm, H. L., Sampson, M. G., & **Bockenhauer, D**. (2020). Diagnoses of uncertain significance: kidney genetics in the 21st century. Nature Reviews Nephrology.
12. Ashton, E., & **Bockenhauer, D**. (2020). Diagnosis of uncertain significance: can next generation sequencing replace the clinician? Kidney International.
13. Govers, L. P., Toka, H. R., Hariri, A., Walsh, S. B., & **Bockenhauer, D**. (2020). Mitochondrial DNA mutations in renal disease: an overview. Pediatric Nephrology. doi:10.1007/s00467-019-04404-6
14. Downie, M. L., & **Bockenhauer, D**. (2019). Swimming with the fishes: delineating tubular transport pathways for magnesium.. Pflugers Arch. doi:10.1007/s00424-019-02286-z
15. Besouw, M. T. P., Kleta, R., & **Bockenhauer, D**. (2019). Bartter and Gitelman syndromes: questions of class. Pediatric Nephrology. doi:10.1007/s00467-019-04371-y
16. Outtandy P, Russell C, Kleta R, **Bockenhauer D**.: Zebrafish as a model for kidney function and disease. Pediatr Nephrol. 2018 Mar 3. doi: 10.1007/s00467-018-3921-7. [Epub ahead of print]
17. **Bockenhauer, D.**, & Kleta, R. (2017). Salt-losing tubulopathies in children: what's new, what's controversial? J Am Soc Nephrol. 2017 Dec 13. pii: ASN.2017060600. doi: 10.1681/ASN.2017060600. [Epub ahead of print]
17. Gupta S, Köttgen A, Hoxha E, Brenchley P, **Bockenhauer D**, Stanescu HC, Kleta R.: Genetics of membranous nephropathy. Nephrol Dial Transplant. 2017 Nov 6. doi: 10.1093/ndt/gfx296. [Epub ahead of print]
19. Kesselheim, A., Ashton, E., & **Bockenhauer, D**. (2017). Potential and pitfalls in the genetic diagnosis of kidney diseases. Clin Kidney J. 2017 Oct;10(5):581-585
20. **Bockenhauer D**. Hyponatremia and cyst growth in neonatal polycystic kidney disease: a case for aquaretics? Pediatr Nephrol. 2017 Feb 13 [Epub ahead of print]
21. **Bockenhauer D**, Bichet DG. Nephrogenic diabetes insipidus. Curr Opin Pediatr. 2017 Apr;29(2):199-205
22. **Bockenhauer D**, Kleta R.: Of dogs and men. Eur J Hum Genet. 2017 Feb;25(2):161
23. Oliveira B, Kleta R, **Bockenhauer D**, Walsh SB: Genetic, Pathophysiological and Clinical Aspects of Nephrocalcinosis. Am J Physiol Renal Physiol. 2016 Dec 1;311(6):F1243-F1252
24. Abdelhadi O, Iancu D, Stanescu H, Kleta R, **Bockenhauer D**: EAST syndrome: Clinical, pathophysiological, and genetic aspects of mutations in KCNJ10. Rare Dis. 2016 Jun 1;4(1):e1195043
25. Wallace D, Lichtarowicz-Krynska E, **Bockenhauer D:** Non-accidental salt poisoning. Arch Dis Child. 2017 Feb;102(2):119-122
26. Viering DH, de Baaij JH, Walsh SB, Kleta R, **Bockenhauer D:** Genetic causes of hypomagnesemia, a clinical overview. Pediatr Nephrol. 2016 May 27. [Epub ahead of print]

27. Bichet DG, **Bockenhauer D:** Genetic forms of nephrogenic diabetes insipidus (NDI): Vasopressin receptor defect (X-linked) and aquaporin defect (autosomal recessive and dominant). Best Pract Res Clin Endocrinol Metab. 2016 Mar;30(2):263-76

28. **Bockenhauer D**, Jaureguiberry G. HNF1B-associated clinical phenotypes: the kidney and beyond. Pediatr Nephrol. 2016 May;31(5):707-14

29. **Bockenhauer D**, Bichet DG: Pathophysiology, diagnosis and management of nephrogenic diabetes insipidus. Nat Rev Nephrol. 2015 Oct;11(10):576-88. doi: 10.1038/nrneph.2015.89

30. Klootwijk ED, Reichold M, Unwin RJ, Kleta R, Warth R, **Bockenhauer D**.: Renal Fanconi syndrome: taking a proximal look at the nephron. Nephrol Dial Transplant. 2015 Sep;30(9):1456-60.

31. **Bockenhauer D**, Zieg J.: Electrolyte disorders. Clin Perinatol. 2014 Sep;41(3):575-90

32. **Bockenhauer D**: Draining the edema: a new role for aquaretics? Pediatr Nephrol. 2014 May;29(5):767-9

33. Kari JA, **Bockenhauer D**, Stanescu H, Gari M, Kleta R, Singh AK: Consanguinity in Saudi Arabia: A Unique Opportunity for Pediatric Kidney Research. Am J Kidney Dis. 2014 Feb;63(2):304-10

34. **Bockenhauer D**: Over- or underfill: not all nephrotic states are created equal. Pediatr Nephrol. 2013 Aug;28(8):1153-6.

35. **Bockenhauer D**, Bichet DG: Inherited secondary nephrogenic diabetes insipidus: Concentrating on humans. Am J Physiol Renal Physiol. 2013 Apr 15;304(8):F1037-42

36. Russell-Eggitt I, **Bockenhauer D**: The blind kidney: disorders affecting kidneys and eyes. Pediatr Nephrol. 2013 Dec;28(12):2255-65

37. Aitkenhead, H and **Bockenhauer D**: The kidney speaks: Interpreting urinary electrolytes, Arch Dis Child, 2011;96:223-227

38. Bandulik, S., Schmidt, K., **Bockenhauer, D.**, Zdebik, A. A., Humberg, E., Kleta, R., Warth, R., Reichold, M. (2011). The salt-wasting phenotype of EAST syndrome, a disease with multifaceted symptoms linked to the KCNJ10 K+ channel. Pflugers Arch 461(4), 423-435

39. Kleta, R., Klootwijk, E., Stanescu, H., **Bockenhauer, D**. (2011). Filtering the genes and sorting the glomerular filter: a new piece in the puzzle? Nephrol Dial Transplant 26(9), 2743-2745

40. Adalat, S., **Bockenhauer, D.**, Ledermann, S. E., Hennekam, R. C., Woolf, A. S. (2010). Renal malformations associated with mutations of developmental genes: messages from the clinic. Pediatr Nephrol 25(11), 2247-2255

41. **Bockenhauer, D.**, Medlar, A. J., Ashton, E., Kleta, R., Lench, N. (2011). Genetic testing in renal disease. Pediatr Nephrol 2012 Jun;27(6):873-83

42. **Bockenhauer, D.**, Hug, M. J., Kleta, R. (2009). Cystic fibrosis, aminoglycoside treatment and acute renal failure: the not so gentle micin. Pediatr Nephrol 24(5), 925-928

43. Camargo, S. M. R., **Bockenhauer D.**, Kleta, R. (2008). Aminoacidurias: Clinical and molecular aspects. Kidney International 73(8), 918-925

44. Kleta, R., **Bockenhauer D**. (2006). Bartter syndromes and other salt-losing tubulopathies. Nephron Physiology 104(2), p73-p80

45. **Bockenhauer, D**. (2001). Ion channels in disease. Curr Opin Pediatr 13(2), 142-149

46. Goldstein, S. A., **Bockenhauer D.**, O'Kelly, I., Zilberberg, N. (2001). Potassium leak channels and the KCNK family of two-P-domain subunits. Nat Rev Neurosci 2(3), 175-184.

47. **Bockenhauer D**, Stanescu H, Bandulik S, Reichold M, Zdebik A, Warth R, Kleta R (2011) EAST-Syndrom: Ein neues Krankheitsbild mit renalem Salzverlust. Nephrologe 2011(6):529–536

Projects

Cliical trial in rare kidney diseases (NDI, dRTA, PKD etc.)

Research into inherited kidney diseases

Memberships

- Belgian Paediatric Nephrology
- Belgian Nephrology Society
- Gesellschaft für Paediatrische Nephrologie (Germany)
- Deutsche Gesellschaft für Nephrologie
- European Society for Paediatric Nephrology
- International Paediatric Nephrology Association
- American Society of Nephrology
- European Renal Association/European Dialysis and Transplant Association

Other Relevant Information