

Rare Kidney Stone Consortium

Book Chapters

1. Edvardsson VO, Palsson R, Sahota A. Adenine Phosphoribosyltransferase Deficiency. In: Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP, eds. *GeneReviews*. Seattle WA: University of Washington, Seattle; 1993. PMID: 22934314
2. Lieske JC, Milliner DS, Beara-Lasic L, Rossetti S. Dent Disease. In: Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP, eds. *GeneReviews*. Seattle WA: University of Washington, Seattle; 1993. PMID: 22876375

Abstracts Presented at Conferences

1. Edvardsson V, Gudbjartsson D, Indridason O, et al. Sequence Variants at the UMOD Gene Associate with Risk of Kidney Stone Disease. American Society of Nephrology Kidney Week. San Diego, CA2009.
2. Edvardsson V, Palsson R, Indridason O, Thorvaldsson S, Stefansson K. Familial Clustering of Kidney Stone Disease in Iceland. Biennial Meeting of The Nordic Society of Nephrology. Helsinki, Finland2009.
3. Indridason O, Gudbjartsson D, Edvardsson V, et al. The Association Between Chronic Kidney Disease and Sequence Variants in the UMOD Gene is Highly Dependent on Age. American Society of Nephrology Kidney Week. San Diego, CA2009.
4. Milliner D, Monico C, Bergstralh E, Lieske J. Urine oxalate variability in primary hyperoxaluria. American Society of Nephrology Kidney Week. San Diego, CA2009.
5. Monico C, Olson J, Bergstralh E, Heilman R, Milliner D. Effect of betaine on urine oxalate in primary hyperoxaluria, type I. American Society of Nephrology (ASN) Kidney Week. Philadelphia, PA2011.
6. Lieske J. How do kidney stones start? Paper presented at: Second International Istanbul Urolithiasis Days; February, 2011; Istanbul, Turkey.
7. Lieske J. What can epidemiology teach us regarding the pathogenesis and consequences of kidney stones? Paper presented at: Second International Istanbul Urolithiasis Days; February, 2011; Istanbul, Turkey.
8. Milliner D. Hyperoxaluria: an update. Paper presented at: Pediatric Nephrology Seminar XXXVIII; March, 2011; Miami Beach, Florida.
9. Lewis M, Sahota A, Ward MD, Goldfarb DS, Holmes E, Want EJ. Characterization of small molecule aggregate components in cystine stones as a screen for candidate stone inhibitors. Paper presented at: American Society for Mass Spectrometry; June, 2011; Denver, CO.
10. Milliner D. Effects of betaine as molecular chaperone in primary hyperoxaluria, type I. Paper presented at: Research on Calculus Kinetics Society; September, 2011; Boston, Massachusetts.
11. Langman C, Vural G, Brooks E. Proteomics in genetic kidney stone disease. Paper presented at: 44th Annual Scientific Meeting of European Society of Paediatric Nephrology (ESPN); September 14-17, 2011; Dubrovnik, Croatia.

12. Goldfarb D. Update on cystinuria: registry and new therapeutic approaches. Paper presented at: Council of Pediatric Nephrology and Urology Meeting; October, 2011; New York.
13. Sahota A, Yang M, Shikhel S, Kathuria N, Lewis M, Ward MD, Goldfarb DS, Tischfield JA. A new pharmacologic approach for cystinuria. Paper presented at: American Society for Human Genetics; October, 2011; Montreal, Canada.
14. Sahota A, Yang M, Shikhel S, Lewis M, Goldfarb DS, Ward MD, Tischfield JA. Tailored inhibition of cystine stone formation as a therapy for cystinuria. Paper presented at: Society for the Study of Inborn Errors of Metabolism (SSIEM); November, 2011; United Kingdom.
15. Sigurdsson B, Thorsteinsdottir M, Eiriksson FF, Edvardsson VO, Palsson R. Rapid method for determination of urinary 2,8-dihydroxyadenine concentration with by UPLC-MS/MS. Paper presented at: Annual Meeting of the European Bioanalysis Forum; November, 2011; Barcelona, Spain.
16. Mattoo A, Modersitzki F, Asplin J, Cohen BH, Grasso M, Goldfarb DS. Clinical validation of a novel assay used for monitoring treatment of patients with cystinuria. Paper presented at: Annual meeting of the American Society of Nephrology; November, 2011; Philadelphia, PA.
17. Chopra B, Amer H, Rodrigo E, Lieske J. Oxalosis after kidney transplantation in bariatric surgery patients: a retrospective analysis. American Society of Nephrology (ASN) Kidney Week. San Diego, CA2012.
18. Edvardsson V. Clinical features and long-term renal outcome in Icelandic patients with APRT deficiency and 2,8-dihydroxyadeninuria. *The 11th Congress of the European Society of Internal Medicine*. Madrid, Spain2012.
19. Eirin A, Irazabal M, Lieske J, Nasr S, Sethi S, Fervenza F. Dent's Disease Presenting as Global Glomerulosclerosis. American Society of Nephrology (ASN) Kidney Week. San Diego, CA2012.
20. Lieske J. Kidney Stones 2012 Overview/ Summary. *American Society of Nephrology Kidney Week*. San Diego, CA2012.
21. Lieske J. Primary Hyperoxaluria: not such a rare disease? *Research on Calculous Kinetics Society Session at the American Urological Association Meeting*. Atlanta, GA2012.
22. Lieske J, Bergstralh E, Mehta R, Langman C, Milliner D. Determinants and outcome of renal calcification in primary hyperoxaluria. American Society of Nephrology (ASN) Kidney Week. San Diego2012.
23. Runolfsson H, Palsson R, Agustsdottir I, Edvardsson V. Clinical Features and Long-Term Renal Outcome in Icelandic Patients with APRT Deficiency and 2,8-Dihydroxyadeninuria. American Society of Nephrology (ASN) Kidney Week. San Diego, CA2012.
24. Edvardsson V. Rare causes of kidney stones and kidney failure: focus on APRT deficiency and 2, 8-dihydroxyadeninuria. Paper presented at: Symposium on Rare Disease, Annual Meeting of the Icelandic Medical Association; January, 2012; Reykjavic, Iceland.
25. Lieske J. Stones from the inside out: what every urologist sees but few nephrologists know about. Paper presented at: Hong Kong Society of Nephrology, Renal Commissioned Training; January, 2012; Hong Kong, People's Republic of China.
26. Goldfarb D. Cysinturia. Paper presented at: 12th International Symposium on Urolithiasis; May, 2012; Ouro Preto, Brazil.

27. Lieske J. Hypertension and Urolithiasis. Paper presented at: 12th International Symposium on Urolithiasis; May, 2012; Ouro Preto, Brazil.
28. Edvardsson VO, Sigurdsson B, Thorsteinsdottir M, Palsson R. Rapid determination of urinary 2,8-dihydroxyadenine with liquid chromatography - electrospray tandem mass spectrometry for monitoring of drug treatment. Paper presented at: The 49th Annual ERA-EDTA Congress; May, 2012; Paris, France.
29. Cohen J, Goldfarb DS, Grasso M. Management of cystinuric patients in a dedicated stone clinic decreases stone events. Paper presented at: Annual Meeting of the American Urological Association; May, 2012; Atlanta, GA.
30. Cohen J, Mattoo A, Modersitzki F, Asplin J, Goldfarb DS, Grasso M. Clinical validation of a novel assay used for monitoring treatment of patients with cystinuria. Paper presented at: Annual Meeting of the American Urological Association; May, 2012; Atlanta, GA.
31. Tosetto E, Casarin A, Cristofaro R, Salviati L, Anglani F. The complexity of the 5'UTR region of CLCN5 gene: ten 5'UTR ends are differentially expressed in the human kidney. Paper presented at: 49th ERA-EDTA Congress; May, 2012; Paris, France.
32. Thorsteinsdottir M, Sigurdsson B, Edvardsson VO, Palsson R. Chemometric optimization of UPLC-MS/MS assay for rapid determination of urinary 2,8-dihydroxyadenine for therapeutic drug monitoring of APRT deficient patients. . Paper presented at: The 60th ASMS Conference on Mass Spectrometry and Allied Topics; May 2012, 2012; Vancouver, BC, Canada.
33. Lieske J. Primary hyperoxaluria: not such a rare disease? Paper presented at: Research on Calculous Kinetics Society Session at the American Urological Association Meeting; May, May 2012; Atlanta, GA.
34. Edvardsson VO, Hardarson S, Palsson R. Renal histopathological findings in patients with 2, 8-Dihydroxyadeninuria. Paper presented at: 49th Annual ERA-EDTA Congress; May 24-27, 2012; Paris, France.
35. Milliner D. Future treatment options/studies. Paper presented at: 10th International Primary Hyperoxaluria Workshop; June, 2012; Bonn, Germany.
36. Thorsteinsdottir M, Sigurdsson B, Eiriksson F, Edvardsson VO, Palsson R. Rapid determination of urinary 2,8-dihydroxyadenine by UPLC-MS/MS for therapeutic drug monitoring of APRT deficient patients. Paper presented at: The XXXIII Meeting of the Nordic Society for Clinical Chemistry; June, 2012; Reykjavik, Iceland.
37. Khouli S, Khandrika L, Meacham R, Olson J, Lieske JC, Milliner D. Culture of hepatocytes from primary hyperoxaluria type I liver. Paper presented at: 10th International Primary Hyperoxaluria Workshop; June, 2012; Bonn, Germany.
38. Knight J, Riedel TJ, Lowther WT, Holmes RP. Measurement of 4-hydroxy-2-oxoglutarate in urine, liver and sera from primary hyperoxaluria type 3 patients Paper presented at: 10th International Primary Hyperoxaluria Workshop; June, 2012; Bonn, Germany.
39. Lorenz E, Dyck PJ, Tracy J, Milliner D. Progressive demyelinating polyradiculoneuropathy due to intraneural oxalate deposition in two patients with type I primary hyperoxaluria (PHI). Paper presented at: 10th International Primary Hyperoxaluria Workshop; June, 2012; Bonn, Germany.

40. Cornell LD, Pamidi N, Nasr SH, Milliner D. Features of recurrent oxalosis in the renal allograft. Paper presented at: 10th International Primary Hyperoxaluria Workshop; June, 2012; Bonn, Germany.
41. Edvardsson V. APRT deficiency and dihydroxyadeninuria: an underrecognized cause of kidney stones and CKD. Paper presented at: Annual Meeting of the ROCK Society; September, 2012; Indianapolis, Indiana.
42. Milliner D. Crystal nephropathies and chameleons. Paper presented at: 8th Uruguayan Congress of Nephrology; September, 2012; Montevideo, Uruguay.
43. Milliner D. Kids and stones and bones. Paper presented at: 8th Uruguayan Congress of Nephrology; September, 2012; Montevideo, Uruguay.
44. Edvardsson VO, Hardarson S, Palsson R. Renal histopathological findings in patients with 2,8-dihydroxyadeninuria. Paper presented at: European Society For Pediatric Nephrology; September, 2012; Krakow, Poland.
45. Edvardsson V, Agustsdottir I, Palsson R. Childhood presentation and renal outcome of APRT deficiency (2,8-Dihydroxyadeninuria) in Iceland. Paper presented at: European Society For Pediatric Nephrology, 45th Annual Meeting; September, 2012; Krakow, Poland.
46. Thorsteinsdottir M, Bragi Sigurdsson B, Eiriksson FF, Edvardsson VO, Palsson R. Chemometric optimization of UPLC-MS/MS assay for evaluation of rare causes of kidney stones and kidney failure. Paper presented at: Waters 3rd Nordic MS Symposium; Sept 11-12, 2012; Jurmala, Latvia.
47. Edvardsson V. Clinical features and long-term renal outcome of Icelandic patients with APRT deficiency and 2,8-dihydroxyadeninuria. Paper presented at: 11th Congress of the European Society of Internal Medicine; October, 2012; Madrid, Spain.
48. Runolfsson H, Edvardsson VO, Hardarson S, Palsson R. Renal histopathological findings in patients with 2,8-dihydroxyadeninuria. Paper presented at: The 11th Congress of the European Society of Internal Medicine; October, 2012; Madrid, Spain.
49. Edvardsson VO, Runolfsson H, Agustsdottir I, Palsson R. Long-term renal outcome in patients with APRT deficiency and 2,8-dihydroxyadeninuria. Paper presented at: Annual Meeting of The American Society of Nephrology; November, 2012; San Diego, CA.
50. Edvardsson V. APRT deficiency. Nordic Society of Nephrology 32nd Biennial Congress. Reykjavik, Iceland 2013.
51. Lieske J. Bariatric Surgery and Nephrolithiasis. Nephrolithiasis: a systemic disorder. Rome, Italy 2013.
52. Lieske J. Dent Disease. Nordic Society of Nephrology 32nd Biennial Congress. Reykjavik, Iceland 2013.
53. Milliner D. Primary Hyperoxaluria. Nordic Society of Nephrology 32nd Biennial Congress. Reykjavik, Iceland 2013.
54. Thorsteinsdottir M. Quantification of Purine Biomarkers by UPLC-MS/MS for Clinical Diagnostic of Rare Kidney Stones and Kidney Failure. 61st ASMS Conference on Mass Spectrometry. Minneapolis, MN 2013.

Journal Articles

1. Hoppe B, Beck BB, Milliner DS. The primary hyperoxalurias. *Kidney Int.* Jun 2009;75(12):1264-1271. PMID: 19225556
2. Mookadam F, Kheir ME, Alharthi M, Moustafa S, Kazmier FJ, McBain R, Mookadam M, Tajik J, Milliner DS. Vascular Involvement in Primary Hyperoxalosis: An Evidence Based Systematic Overview Over a Fifty Year Span. *Nephro Urol Mon.* 2010;02(03):390-396.
3. Nasr SH, Sethi S, Cornell LD, Milliner DS, Boelkins M, Broviac J, Fidler ME. Crystalline nephropathy due to 2,8-dihydroxyadeninuria: an under-recognized cause of irreversible renal failure. *Nephrol. Dial. Transplant.* Jun 2010;25(6):1909-1915. PMID: 0064951
4. Chillaron J, Font-Llitjos M, Fort J, Zorzano A, Goldfarb DS, Nunes V, Palacin M. Pathophysiology and treatment of cystinuria. *Nature reviews. Nephrology.* Jul 2010;6(7):424-434. PMID: 20517292
5. Belostotsky R, Seboun E, Idelson GH, Milliner DS, Becker-Cohen R, Rinat C, Monico CG, Feinstein S, Ben-Shalom E, Magen D, Weissman I, Charon C, Frishberg Y. Mutations in DHDP5L are responsible for primary hyperoxaluria type III. *Am. J. Hum. Genet.* Sep 10 2010;87(3):392-399. PMID: 20797690, PMCID: PMC2933339
6. Rimer JD, An Z, Zhu Z, Lee MH, Goldfarb DS, Wesson JA, Ward MD. Crystal growth inhibitors for the prevention of L-cystine kidney stones through molecular design. *Science.* Oct 15 2010;330(6002):337-341. PMID: 20947757
7. Bergstralh EJ, Monico CG, Lieske JC, Herges RM, Langman CB, Hoppe B, Milliner DS. Transplantation outcomes in primary hyperoxaluria. *Am. J. Transplant.* Nov 2010;10(11):2493-2501. PMID: 20849551, PMCID: PMC2965313
8. Mookadam F, Smith T, Jiamsripong P, Moustafa SE, Monico CG, Lieske JC, Milliner DS. Cardiac abnormalities in primary hyperoxaluria. *Circ. J.* Nov 2010;74(11):2403-2409. PMID: 20921818
9. Anglani F, Del Prete D, Peruzzi L. [Participation in the International Dent's disease registry: clinical spin-offs and research prospects]. *Giornale italiano di nefrologia : organo ufficiale della Societa italiana di nefrologia.* Jan-Feb 2011;28(1):7-8. PMID: 21341234
10. Lee HJ, Jeong SJ, Lee EO, Bae H, Lieske JC, Kim SH. 1,2,3,4,6-Penta-O-galloyl-beta-D-glucose reduces renal crystallization and oxidative stress in a hyperoxaluric rat model. *Kidney Int.* Mar 2011;79(5):538-545. PMID: 1085110
11. Murphy S, Reilly MM. Amyloid neuropathies. *Advances in Clinical Neuroscience and Rehabilitation.* March/April 2011;11(1):16-19.
12. Coe FL, Evan A, Worcester E. Pathophysiology-based treatment of idiopathic calcium kidney stones. *Clin. J. Am. Soc. Nephrol.* Aug 2011;6(8):2083-2092. PMID: 21825103
13. Rule AD, Krambeck AE, Lieske JC. Chronic kidney disease in kidney stone formers. *Clin. J. Am. Soc. Nephrol.* Aug 2011;6(8):2069-2075. PMID: 21784825, PMCID: PMC3156433
14. Goldfarb DS. Potential pharmacologic treatments for cystinuria and for calcium stones associated with hyperuricosuria. *Clin. J. Am. Soc. Nephrol.* Aug 2011;6(8):2093-2097. PMID: 21757641, PMCID: PMC3156434
15. Monico CG, Rossetti S, Belostotsky R, Cogal AG, Herges RM, Seide BM, Olson JB, Bergstrahl EJ, Williams HJ, Haley WE, Frishberg Y, Milliner DS. Primary hyperoxaluria type III gene HOGA1

(formerly DHDSL) as a possible risk factor for idiopathic calcium oxalate urolithiasis. *Clin. J. Am. Soc. Nephrol.* Sep 2011;6(9):2289-2295. PMID: 21896830, PMCID: PMC3358997

16. Hoppe B, Groothoff JW, Hulton SA, Cochat P, Niaudet P, Kemper MJ, Deschenes G, Unwin R, Milliner D. Efficacy and safety of Oxalobacter formigenes to reduce urinary oxalate in primary hyperoxaluria. *Nephrol. Dial. Transplant.* Nov 2011;26(11):3609-3615. PMID: 21460356
17. Lee HJ, Jeong SJ, Park MN, Linnes M, Han HJ, Kim JH, Lieske JC, Kim SH. Gallotannin suppresses calcium oxalate crystal binding and oxalate-induced oxidative stress in renal epithelial cells. *Biol. Pharm. Bull.* 2012;35(4):539-544. PMID: 22466558
18. Beara-Lasic L, Edvardsson V, Palsson R, Lieske J, Goldfarb D, Milliner D. Genetic Causes of Kidney Stones and Kidney Failure. *Clinical Reviews in Bone and Mineral Metabolism.* 2012;10(1):2-18.
19. de Cogain M, Krambeck AE, Rule AD, Li X, Bergstralh EJ, Gettman MT, Lieske JC. Shock wave lithotripsy and diabetes mellitus: a population-based cohort study. *Urology.* Feb 2012;79(2):298-302. PMID: 22088569, PMCID: PMC3274621
20. Monico CG, Milliner DS. Genetic determinants of urolithiasis. *Nature reviews. Nephrology.* Mar 2012;8(3):151-162. PMID: 22183508
21. Schinstock CA, Semret MH, Wagner SJ, et al. Urinalysis is more specific and urinary neutrophil gelatinase-associated lipocalin is more sensitive for early detection of acute kidney injury. *Nephrol. Dial. Transplant.* May 2013;28(5):1175-1185. PMID: 22529161
22. Rule AD, Sasiwimonphan K, Lieske JC, Keddis MT, Torres VE, Vrtiska TJ. Characteristics of renal cystic and solid lesions based on contrast-enhanced computed tomography of potential kidney donors. *Am. J. Kidney Dis.* May 2012;59(5):611-618. PMID: 22398108, PMCID: PMC3328591
23. Dwyer ME, Krambeck AE, Bergstralh EJ, Milliner DS, Lieske JC, Rule AD. Temporal trends in incidence of kidney stones among children: a 25-year population based study. *J. Urol.* Jul 2012;188(1):247-252. PMID: 22595060, PMCID: PMC3482509
24. Pang R, Linnes MP, O'Connor HM, Li X, Bergstralh E, Lieske JC. Controlled metabolic diet reduces calcium oxalate supersaturation but not oxalate excretion after bariatric surgery. *Urology.* Aug 2012;80(2):250-254. PMID: 22554593, PMCID: PMC3411876
25. El-Zoghby ZM, Lieske JC, Foley RN, Bergstralh EJ, Li X, Melton LJ, 3rd, Krambeck AE, Rule AD. Urolithiasis and the Risk of ESRD. *Clin. J. Am. Soc. Nephrol.* Sep 2012;7(9):1409-1415. PMID: 22745275, PMCID: PMC3430957
26. Riedel TJ, Knight J, Murray MS, Milliner DS, Holmes RP, Lowther WT. 4-Hydroxy-2-oxoglutarate aldolase inactivity in primary hyperoxaluria type 3 and glyoxylate reductase inhibition. *Biochim. Biophys. Acta.* Oct 2012;1822(10):1544-1552. PMID: 22771891, PMCID: PMC3418427
27. Edvardsson VO, Indridason OS, Haraldsson G, Kjartansson O, Palsson R. Temporal trends in the incidence of kidney stone disease. *Kidney Int.* Jan 2013;83(1):146-152. PMID: 22992468
28. Lorenz EC, Michet CJ, Milliner DS, Lieske JC. Update on oxalate crystal disease. *Curr. Rheumatol. Rep.* Jul 2013;15(7):340. PMID: 23666469, PMCID: PMC3710657
29. Sumorok N, Goldfarb DS. Update on cystinuria. *Curr. Opin. Nephrol. Hypertens.* Jul 2013;22(4):427-431. PMID: 23666417

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35. Modersitzki F, Pizzi L, Grasso M, Goldfarb DS. Health-related quality of life (HRQoL) in cystine compared with non-cystine stone formers. *Urolithiasis.* Feb 2014;42(1):53-60. PMID: 24253538
36. Tang X, Lieske JC. Acute and chronic kidney injury in nephrolithiasis. *Curr. Opin. Nephrol. Hypertens.* Jul 2014;23(4):385-390. PMID: 24848936, PMCID: PMC4096690
37. Meeusen JW, Lieske JC. Looking for a better creatinine. *Clin. Chem.* Aug 2014;60(8):1036-1039. PMID: 24573605
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41. Ketha H, Singh RJ, Grebe SK, et al. Altered Calcium and Vitamin D Homeostasis in First-Time Calcium Kidney Stone-Formers. *PLoS ONE.* 2015;10(9):e0137350. PMID: 26332888, PMCID: PMC4558059
42. Sigurjonsdottir VK, Runolfsdottir HL, Indridason OS, Palsson R, Edvardsson VO. Impact of nephrolithiasis on kidney function. *BMC Nephrol.* 2015;16:149. PMID: 26316205, PMCID: PMC4551564
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