

Dent Disease eNewsletter December 2014



Dear Colleagues,

We're excited to share ongoing activities of the Rare Kidney Stone Consortium (RKSC). Many of you have contacted us in the past and know that we are a resource for health care providers and patients. Our Dent Disease Program is part of the NIH funded RKSC of investigators dedicated to the study of four rare inherited kidney stone diseases: Dent disease, Cystinuria, APRT deficiency, and primary hyperoxaluria, as well as enteric hyperoxaluria. Recently the RKSC was funded for a second 5 year cycle by the NIH. Please visit our RKSC web sites at <http://www.rarediseasesnetwork.org/RKSC/> and www.rarekidneystones.org/, as well as our Dent Disease page on Facebook: <http://www.facebook.com/DentDisease>

Below is a list of our currently active protocols. Please contact us for more information and/or if you have patients that might be interested.

Dent Disease Registry

The RKSC maintains a secure Dent Disease Registry to accumulate important data on Dent disease patients including disease manifestations and responses to treatments. Currently there are 131 patients in the Registry. RKSC staff are available to assist with data entry, or to help local centers with data entry (if desired).

Screening for Dent Disease Mutations

The RKSC continues to offer genetic testing on a research basis for Dent Disease (types I and II). This testing is available to all suspected patients or carriers who meet our inclusion criteria that indicate such testing is warranted. Please contact the RKSC Study Coordinators for additional information: rarekidneystones@mayo.edu or 800-270-4637. RKSC staff and physicians are available to discuss testing options, interpretation of test results, and for questions regarding clinical management.

Dent Biobank

The RKSC maintains a biobank for future studies that includes blood and urine samples from Dent patients and carriers.

Systematic review of clinical biopsies of Dent Disease patients

Many patients with Dent disease develop chronic kidney disease. The exact reasons and kidney pathology are not entirely defined. In this protocol we are reviewing kidney biopsy reports and slides (if available) from patients that have had a kidney biopsy during the course of their care in order to understand the kidney manifestations better. Currently we have 21 patients enrolled. Please contact us if you are interested in collaborating on this project.

Role of phosphorus and FGF23 in Patients with Dent Disease

Many patients with Dent disease have hypercalciuria and develop nephrocalcinosis as well as significant bone disease/growth delay. The underlying reasons for the hypercalciuria are not clearly understood, and the ideal treatment strategy is not yet worked out. In this protocol we are

systematically examining the hormonal systems that regulate calcium and phosphorus metabolism in a group of Dent disease patients, as well as a second group without Dent disease but with calcium oxalate kidney stones. Patients will be studied before and after two weeks of treatment with neutral phosphorus. Please contact us if you would like further details or have Dent disease or kidney stone patients that are potentially interested in participating in this trial.

Dent disease prospective cohort study (awaiting IRB approval).

An important new study in our new grant cycle will be to develop a prospective cohort of patients for a standardized yearly follow up. Patients will have history, blood and urine tests... In this manner we will be able to have specific data elements and bio specimens available for detailed studies regarding the pathogenesis of the disease. We will be recruiting patients with Dent disease type I and 2, Lowe syndrome, and Dent carriers. Please contact us if you have any patients that are interested in participating.

Recent Publications

The RKSC recently published a paper reviewing rare kidney stone diseases in *Pediatric Nephrology*: "Hereditary causes of kidney stones and chronic kidney disease", currently online at: <http://www.ncbi.nlm.nih.gov/pubmed/23334384>. This publication also includes diagnostic algorithms for each of the four diseases (Edvardsson VO, Goldfarb DS, Lieske JC, *et al.* Hereditary causes of kidney stones and chronic kidney disease. *Pediatric nephrology* 2013). The RKSC investigators also recently authored the section in GeneReviews on Dent Disease (Lieske JC, Milliner DS, Beara-Lasic L, Harris P, Hopp K, Cogal A, Mattison K. Dent Disease. 2012 Aug 09 [updated 2014 Sep 25]. In: Pagon RA, Adam MP, Ardinger HH, Bird TD, Dolan CR, Fong CT, Smith RJH, Stephens K, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2014. Available from <http://www.ncbi.nlm.nih.gov/books/NBK99494/> PubMed PMID: 22876375.)

Training Program and Pilots

A key mission of the RKSC is the training of young investigators in rare disease research, and supporting pilot projects to address key research questions in rare kidney stone disease. We expect to have a request for proposals for both programs in early 2015. Please contact us for further details.



DENT DISEASE

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WELCOME TO THE DENT DISEASE FOUNDATION

The Dent Disease Foundation's purposes are:

- To raise funds to support research into the causes of the disease so we may halt its progression and find a cure;
- To educate patients, the medical community and the public; and to promote communication among patients, their families and the medical community.

The Dent Disease Foundation, a non-profit 501c organization provides peer support and is an excellent resource for patients and their families (<http://www.dentdisease.com>). Together with the

RKSC, they hosted 2 patient and family workshops in Rochester, MN (June 2013) and Chicago, IL (June, 2014). Plans are underway for the 3rd patient meeting in mid 2015 in New York, possibly including a one-day scientific program. Please contact Catherine Forrest if you have patients or are interested in working with the Dent Disease Foundation (caforrest@dentdisease.com).

The RKSC Awareness Walk, June 2014.



We would like to thank everyone who has worked with the RKSC in furthering Dent Disease research and continuing to strive to provide better treatment for our Dent disease patients. We look forward to all future collaborations and inquiries and we are here to help!

Rare Kidney Stone Consortium

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