Dear Registry Participant,

Welcome to the first Cystinuria Registry Newsletter. We would like to share with you the following topics.

2010 was a very successful year in our Rare Kidney Stone Consortium and the Cystinuria registry in particular. Please find a summary of these exciting times and a brief outlook for 2011 below. We continue with our effort to enroll participants into the registry.

You are all playing an active role in this registry and your support is deeply appreciated. Do not hesitate to contact us at cystinuria@nyumc.org with all cystinuria-related questions.

- Frank Modersitzki, MPH

We would also like highlight our publication in the journal Science. The development of a compound that inhibits the growth of cystine crystals was first presented by Dr. Michael Ward, Chair of the Chemistry Department at NYU, at the ICF symposium at NYU Medical Center in summer 2010. The corresponding article followed in fall 2010. This compound is now being tested in mice by Dr. Amrik Sahota at Rutgers. We are very excited about the preliminary results. We are also working on the final report of our Quality of Life survey and hope to publish this paper in early 2011. We presented a poster of additional results at the American Society of Nephrology in Denver in November, 2010 (see picture).

Activation of Contact and Disease Registry

As you all know, we went live with the Contact and Disease registries in late October of 2010. Cystinuria is one of four rare kidney stone diseases that are collaborating in this consortium. (The others are primary hyperoxaluria, APRT deficiency and Dent Disease). To date, almost 180 people signed up for the contact registry and all of you should have received a consent form for the “disease” registry by email. The first 50 subjects have now returned their consent forms. We have collected the first sets of patient charts and the data entry into the disease registry is proceeding.

- Frank Modersitzki, MPH

Dr. David Goldfarb and Frank Modersitzki in front of their poster presentation on quality of life in cystinuria at the American Society of Nephrology (ASN) in Denver, November, 2010

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Patient Advocacy Groups

Cystinuria Support Network - Kathryn Jewell's Report

I was born Kathryn Jewell in November 1959 in California: a healthy child. At the age of about 7 cystinuria was discovered. I had been anemic, then had severe right lower quadrant pain. Assuming appendicitis they were prepping me for surgery, and found kidney stones on the x-ray. By the age of 18 I had had multiple invasive surgeries and had a partial nephrectomy because my kidney was found to be packed full of stones again. Much of my childhood was spent in pain, and I was in and out of hospitals I struggled to find out more information on cystinuria, as there was so little out there. In Houston Texas, in the early 80’s, I had a lithotripsy done before it was approved by the FDA. It was so painful, an intense procedure, but a much better option than the invasive surgical procedures that I had already had 4 of, all on the same kidney. The doctors told me that surgery on that kidney was no longer an option because of so much scar tissue. Finally the internet became available to me in the the late 1990’s and I got in touch with Matt Lewis, and also the Cystinuria Support Network, and finally, my hero, Dr David Goldfarb. From that moment on, I had a resource, and my life changed forever. I knew that I had to know more than my doctors about the disease, and educate them. Most were open, a few were not, yet I had the information to guide them if they used it. I moved 36 times, being a military brat, then an oil field wife, and later a traveling sonographer. Moving became an obstruction to my care. My medical records were a nightmare. As of 2010, I sadly had to give up my career as a sonographer because of cystinuria, and go on disability. It was a long hard fight because of the ignorance people have of this disease. When I was asked to become involved in the Rare Kidney Stone Consortium (RKSC) I was honored. I will offer up myself, in any way, for research. This was one way that I could hopefully help others with the disease. It has been an honor to listen in on phone calls to some of the best doctors, researchers, and staff that are involved with the RKSC. Now that all the protocols for the grants, research, and most of the guideline paperwork is complete for RKSC, I am really looking forward to hearing about the upcoming research, and hopefully being involved in that on a more personal level.

- Kathryn Jewell

For years, the ICF has connected the international cystinuric population to the world’s leading experts in cystine stone prevention, treatment, and research. Now, as a member of the Rare Kidney Stone Consortium, we are taking the interaction between patients, physicians and researchers to an unprecedented level of scope and sophistication.

The direct participation of the cystinuric community is fundamental to the research goals of the Consortium. The ICF therefore provides the key link necessary for the enrolment of willing participants and the dissemination of study findings back to the patient community.

Status contact and disease registry

Currently we have more than 180 patients with cystinuria signed up for the “contact” registry. Approximately one third of these patients returned the hard copy of the consent form and will be entered into the “disease” registry. This is an ongoing process. If you have not returned your signed consent form, please do so. We have a new version in place, (version date: 3-Mar-2011), please use this version only. In case you need a new hard copy of the consent form with a postage-paid envelope, feel free to request one at cystinuria@nyumc.org.
First Cystinuria Awareness Day

On June 18, 2011 the International Cystinuria Foundation kicked off its first annual Walk, Run, and Bike Ride to raise awareness and donations for cystinuria. ICF founder and activist, and former president, George Brown provided much of the inspiration for having the event. Among many cities, Brooklyn, NY was one of them.

Hosted by Lisa Litwin on behalf of the ICF, the Brooklyn event was a 2.5 mile event at the Shore Rd. Promenade in Bensonhurst, Brooklyn. Set alongside the Atlantic Ocean with the beautiful Verrazano Narrows Bridge in the distance, it was a perfect day for the Event (see picture). Many friends and relatives also participated.

Contact Dr. Goldfarb (dsgold@verizon.net) if you are interested in getting one of the beautiful ICF bike jerseys, still available. As a result of these events, the ICF successfully raised awareness and more donations than ever, funds that will go to the ICF for further research of cystinuria. The ICF will be holding these events next year as well so please keep an eye on the ICF website (www.cystinuria.org) for details.

You can also follow the International Cystinuria Foundation on Facebook for updates and event details, and speak with other individuals globally that have been affected by cystinuria. We would like to thank all of the individuals that were able to attend the events, spread awareness, and make generous donations. These selfless acts of kindness are greatly appreciated by people faced with cystinuria.

- Lisa Litwin, July 2011

Internet Resources

NYU School of Medicine, Division of Nephrology - main site for the cystinuria registry
http://medicine.med.nyu.edu/nephrology/research/current/cystinuriaregistry -

Cystinuria Support Network, active Yahoo email group
http://www.cystinuria.com
http://health.groups.yahoo.com/group/cystinuria-support-network/

International Cystinuria Foundation
http://www.cystinuria.org

Rare Kidney Stone Consortium, main consortium web site
http://www.rarekidneystones.org

Calendar

November 8 - 13, 2011
ASN Philadelphia

October 8, 2011
ICF Walk Run Bike Ride in Corvallis, OR

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