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2011 Cystinosis Research Network Family Conference in San Francisco a Great Success!



The 2011 Cystinosis Research Network (CRN) Family Conference was held July 14 - 16 in San Francisco, California at the Hotel Nikko. Approximately 250 people were in attendance, including 55 cystinosis families and 50 clinicians, researchers and professionals. Thirteen families were in attendance who were newly diagnosed or who had never attended a conference. Twenty-four states and ten countries were represented. This year's theme, "Bridge to the Future", was evident in the special emphasis placed on the needs of teens and adults with cystinosis, as well as the encouraging research reports which were shared.



Children and adults with cystinosis at the 2011 Cystinosis Research Network Family Conference

With the support of our generous corporate sponsors Sigma-Tau Pharmaceuticals, Raptor Pharmaceuticals, C.H. Robinson Worldwide, Inc., Orphan Europe, Ferrandino and Son, Inc., Maccarone Plumbing, Inc., Pav-Lak Contracting, Inc., as well as the contributions of 15 families, CRN was able to provide three days of programming which was aimed at both families and professionals. Wednesday began with the CRN Board of Directors meeting, a rare opportunity for our hardworking board to meet in person, rather than via conference call as is the norm.

Conference registration began the day on Thursday. The CRN Scientific Review Board met to review the projects submitted through this year's call for proposals. Several new grants were funded, details about which can be found in the research section of this newsletter.

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Message from the President

Christy Greeley

By Christy Greeley*"If not now, when?"*

This quote has followed me around lately. I hear it during political debates on television and inserted into musical lyrics. Most recently, I received a paperweight in the mail with the saying inscribed on it from a fellow board member. She had recently attended the Genetic Alliance 25th Anniversary Gala in Washington, D.C. with others from CRN. Dr. Francis Collins, Director of the National Institutes of Health, played a song on his guitar to wrap up the gala and ended **the performance with the words, "If not now, when?"** She saw the paperweight in the airport on her trip home and took it as a sign.

So I looked into the origin of the quote and found it is attributed to Hillel the Elder, a famous Jewish religious leader and one of the most important figures in Judaic history. The full quote actually reads:

"If I am not for myself, who will be? And when I am for myself, what am 'I'? And if not now, when?"

Perfect. To me, this is the essence of our mission and summarizes CRN's three areas of concentration, Education and Awareness, Family Support, and Research.

"If I am not for myself, who will be?"

Self-advocacy is at the heart of CRN's increased efforts to support and meet the needs of our community. Our children are growing into teens, then productive adults. Our strategy has been to assess their needs and meet them in advance, involving them every step of the way. Thus, the CRN Adult Care Excellence Initiative was formed. It included physicians, parents of teens and young adults with cystinosis, and individuals with cystinosis. They worked for a year under the direction of our advisory board social worker Maya Doyle to produce a transition guide for teens, young adults and their parents. The guide was debuted at the CRN Family Conference in July and can be accessed on the CRN website at <http://www.cystinosis.org/filemanager/file/Resources/Transitioning/CRNtransit06152011.pdf>

The workgroup has turned its focus this year to producing a comprehensive guide for parents and physicians for use in the diagnosis and management of cystinosis. Education and awareness efforts also continue with expanded information available on the website and other channels, active participation on the National Organization of Rare Diseases board of directors and with the Genetic Alliance, and participation and exhibits at professional meetings such as the American Society of Nephrology meeting this November in Philadelphia.



Dave, Jack, Christy, and Alex Greeley at Crater Lake, OR

*denotes President Emeritus

"And when I am for myself, what am I?"

Family support and community building is essential to individual and family long term health and success. We believe that connecting newly diagnosed families with those who have walked in their shoes, providing regional family gatherings and family conferences, and facilitating virtual opportunities for parents, teens and adults with cystinosis to communicate and bond together are the best way to ensure that our message of education, self-advocacy and empowerment are relayed. **This summer's CRN Family Conference, which took place in July in San Francisco, California, provided just that platform.** One especially powerful example from the conference comes to mind. A Patient Panel session took place which featured seven teens and young adults with cystinosis who shared their experience with the disease with attendees, impressing everyone in attendance with their candor, clearly giving of themselves in order to enrich the community. We are blessed with an incredibly motivated, caring and active group of parents, teens and young adults, and clinicians and physicians. CRN continues to seek out new ways to expand on our family support activities.

"And if not now, when?"

Here is our call to action. Action in funding new research grants to expand upon our understanding of the disease, to train new clinicians and researchers, and to hasten better treatment. CRN is funding three new research grants this year totalling over \$300,000, which pushes CRN historical research funding total well over \$3 million. One of these studies represents a joint funding effort between CRN and Cystinosis Foundation Ireland, an innovative collaborative effort to address research issues which have an interest for both groups. In addition, CRN has added several new members to our Scientific Review Board, Medical Advisory Committee and Board of Directors, demonstrating increased involvement from new physicians in the field of cystinosis and well as new families **who have decided to dedicate themselves to service to CRN and the community.** You can read more about CRN's research funding and our new board members in this edition of the newsletter.

We are called to action both locally here in the U.S. as well as globally. CRN is sponsored a Cystinosis Symposium during the Latin American Pediatric Nephrology Association meeting (ALANEPE 2011) in Sao Paulo, Brazil in October. We feel it is our responsibility to assist in the education of physicians all over world so that diagnoses are made more quickly and appropriate treatment can begin for patients in their home countries. We are honored to work with Dr. Vera Koch in the organization of the meeting and feel that international outreach and support of the global cystinosis community is an important part of our mission as we share the knowledge and expertise we are fortunate to have access to.

My family had an "If not now, when?" moment ourselves this July. We moved to Chicago from Portland, Oregon 11 years ago. We lived in a suburb of Portland for three years in the late 1990's – in fact, Jack was born there 9 months before we moved to Chicago. We decided to take the opportunity provided by our attendance at the CRN Family Conference in San Francisco to take another week's vacation and drive up the coast after the meeting to Portland, visiting old friends and a city we had loved living in. In planning the trip, we realized we could stop at one of the sites we had always meant to visit and never gotten around to, Crater Lake, Oregon, the deepest lake in the U.S., situated at nearly 7,000 feet elevation and formed thousands of years ago as the result **of the collapse of a volcano. It was a few very scenic hours' drive off I-5** and would increase our drive that day from about 11 hours to 15, but we figured, when will we ever be driving through south central Oregon again? So we went, and were so thrilled that we had seized the opportunity. The family photo included here was taken as we arrived at Crater Lake, it was breathtaking, a great memory from a great summer vacation.

I encourage you all to take advantage of opportunities as they arise, both in your personal lives as well as within the cystinosis community. We are all too aware of the twists and turns life can take us on -- we are fortunate to be on this road together. I wish you all a wonderful holiday season and a joyous and prosperous 2012!

Warmest Regards,

Christy Greeley

CRN President and Executive Director



Jack, Christy, and Alex Greeley at the 2011 CRN Family Conference

2011 CRN Family Conference in San Francisco a Great Success! (cont.)

Continued from page 1



The Britt and Jondle family

Thursday night really kicked off the conference with a welcome reception hosted by the Jondle and Jordan families, whose hard work made this year's conference in San Francisco possible. Everyone was incredibly moved by both families' heartfelt welcomes to the group -- it really provided a wonderful way for everyone to get reacquainted, re-new old friendships and meet the new families and professionals in attendance.

Highlights of Friday's program included a presentation by Cindy Abbott, a rare disease advocate whose adventures climbing Mt. Everest in spite of her rare disease provided an inspirational kick off for the day. To complete the morning, Dr. Gahl gave an insightful overview of cystinosis and Cheri Friend led the group in a family introduction session. Friday afternoon began with our largest ever Poster Session, which included 25 posters from researchers and industry and advocacy representatives. We also heard updates from the CRN National Institutes of Health Clinical Fellow Dr. Nesterova, Dr. Cherqui on the gene therapy consortium, and representatives from Raptor and Sigma

Tau on the progress of the extended release cystagon and the cysteamine eye drops. The day ended with the annual medical panel, where questions from the audience were answered by 24 cystinosis experts. It was indeed awe-inspiring to see visual confirmation of the wealth of dedicated physicians and researchers who make it a priority to stay in touch with our community and communicate their knowledge and advice.



The Jordan Family

One of Saturday's highlights was the unveiling of the "Building Bridges to the Future" cystinosis transitioning guide. This guide, which was distributed at the meeting and is available on the CRN website, resulted from over a year of work by the CRN Adult Care Excellence Initiative. We urge every family to reference this groundbreaking guide, spearheaded by CRN advisory board member Maya Doyle. Maya's research into transitioning issues in the cystinosis community continues, informed at this meeting by a variety of focus groups. On this day we also heard from the National Organization for Rare Disorders and from cystinosis groups from around the world. The afternoon included workshops sessions geared towards specific topics during which families had the opportunity to interact directly with the experts on topics like childhood issues, transitioning issues, neurology and educational issues, adult issues and transplantation. At the

end of the day, the second "Above and Beyond Award" was presented to Dr. Jerry Schneider for his extraordinary contributions to the cystinosis community. Finally, the highlight of the entire conference may have been the farewell dinner dance held that night, where we were able to spend time together as a cystinosis family...and dance a bit with our favorite doctors!

Childcare this year was exceptional, and included a teen lounge, swimming, arts and crafts, games, face painting and wonderful activities such as a flautist who gave the children music lessons and a Zumba dance instructor who led a great workout for everyone.

Sarah Dawson Photography generously donated her time to photograph the entire conference. Her beautiful photos can be viewed on the CRN website and throughout this newsletter.

We look forward to meeting again in July 2013 in Washington, D.C. Please plan to join us

CRN Founder Colleen Hammond Honored at Genetic Alliance 25th Anniversary Celebration



CRN Board Members Marybeth Kruppenacker, Colleen Hammond, and Frankie McGinnis at the Genetic Alliance 25th Anniversary Celebration

For 25 years the Genetic Alliance has been one of the leaders in health care advocacy. On September 22, 2011 in Washington D.C., one of our own, Colleen Hammond was recognized as being one of 100 **“Innovators of the last 25 year”** at the Genetic Alliance 25th Anniversary Celebration.

The event was held at the National Geographic Society Museum in Washington D.C. The Cystinosis Research Network was represented by Colleen Hammond, Frankie McGinnis, and Marybeth Kruppenacker. Colleen was in good company as some of the other honorees were Dr. Francis Collins, Director of the National Institutes of Health and Dr. Stephen Groft, Director of the Office of Rare Disease Research. The evening was a wonderful opportunity to speak with other innovators and network with some of the

finest individuals in the fields of advocacy, research, and industry as well as hear from people who have done remarkable things!

Colleen was recognized as an “innovator” because of her passion as a mother who wanted the best possible life for her son. Without a strong advocacy group she knew that he might not reach his full potential. So she (along with her husband Jack) founded Cystinosis Research Network in 1996. Colleen believes that every day holds the possibility of miracles, and she will not stop in her mission to find better treatments for cystinosis, not just for her son Shea, but for the entire cystinosis community.

To be an “innovator” one must have tremendous courage, and Colleen certainly does! Those within the cystinosis community and those who know Colleen personally, are well aware that not only is she an incredible wife, mother, daughter, sister, cousin and friend, but she is fierce in her belief that it takes a team in order to make progress toward better lives for individuals with cystinosis. CRN congratulates Colleen on her well-deserved recognition!



Colleen Hammond, Marybeth Kruppenacker, NIH Director Dr. Francis Collins, and Frankie McGinnis



Colleen Hammond, Frankie McGinnis, Office of Rare Diseases Director Dr. Stephen Groft, and Marybeth Kruppenacker

CRN Welcomes Three New Board Members

CRN is pleased to welcome three new board members to our Board of Directors. Rachel DeLomba, Terri Schleuder, and Lynn Thomas all have children with cystinosis and have been active in the cystinosis community, sharing their perspectives of living with cystinosis. Lynn Thomas is a returning board member. We are excited about the knowledge and experience these three ladies will bring to the board. We would also like to thank Lorna Smith and Jill Morrill for their years of service on the board.



Rachel DeLomba

Rachel lives in Illinois and is a stay at home mom to three girls – Anjie, Nelly, and Maya. Anjie and Nelly are both 13 years old and have Cystinosis. Both girls received kidney transplants in December, 2007, from the same donor. Rachel is currently attending Western Governors University where she is pursuing a bachelor's degree in Interdisciplinary Studies K – 8.



Terri Schleuder

Terri and husband Carl live in Novi, Michigan. They have three grown sons, Chris, Eric, and Steve (age 23) with Cystinosis. Terri received a BSN degree from Indiana University in 1978. For the last ten years she has worked in the Novi Community Schools with special education students. Terri and Steve attended their first Cystinosis Conference in 1989, three weeks after his diagnosis at 18 months of age. She and husband Carl hosted the 1998 Cystinosis Conference held in Ann Arbor, Michigan. Since the beginning of their journey Terri has always found the Cystinosis Community a source of support and friendship. She looks forward to working with CRN as a board member. For fun Terri enjoys writing, reading and her six year old grandson, Aidan.



Lynn Thomas

Lynn Thomas has a Bachelors degree in Management Information Systems, a Bachelor of Science in Business, and a Bachelors degree in Accounting from Geneva College in Beaver Falls, PA. During her 25 plus years in the Computer Industry she managed individual computers to large Data Centers. She was involved in installing the earliest of local area networks (LANs) and wide area networks (WANs) for companies like General Electric, Kodak, Ford, General Motors, ALCOA, and many others. Building the infrastructure of what is now known as the **world wide web**. In the health industry she worked as a security specialist to develop and implement all levels of security to insure HIPAA Compliance. She went on to work as a Project Manager and consultant in a variety of IT environments. Her son Garrett is 15 and has Cystinosis. He has been active in research projects throughout the years, and is currently enrolled in the RP103 study. She has previously served on the Board of Directors for CRN working with the Family Support Committee. Returning to the board she is eager to continue the ground breaking work and spreading the hope we have in the future.

Dr. William A. Gahl Receives Prestigious Honor



2011 SCIENCE AND ENVIRONMENT
MEDAL RECIPIENT

WILLIAM A. GAHL
Director, Undiagnosed Diseases Program
National Institutes of Health

The Cystinosis Research Network is bursting with pride at the announcement that "one of our own", Dr. William A. Gahl, is a 2011 recipient of the *Samuel J. Heyman Service to America Medals*. This medal recognizes a federal employee for a significant contribution to the nation in activities related to science and environment, and it is acknowledged as one of the most prestigious awards dedicated to celebrating America's civil servants.

All of us at CRN extend our most sincere and heartfelt congratulations to Dr. Gahl; we know, only too well, how deserving he is of this honor.

Dr. Gahl is recognized as the founding director of the Undiagnosed Diseases Program at the National Institutes of Health (NIH). This program brings together a unique combination of elite medical specialists, researchers and federal resources to unravel baffling, unrecognized illnesses and, in so doing, develop new and innovative approaches for investigating, understanding and diagnosing rare diseases. Dr. Gahl's devotion to the study of rare diseases has likely evolved through his long-standing research interest in the rare genetic disorder Cystinosis

Dr. Gahl is Clinical Director of the National Human Genome Research Institute, Senior Investigator of the Medical Genetics Branch, and Head of the Human Biochemical Genetics Section. He is also the Chairman of CRN's Scientific Review Board. We are so very fortunate!

To read more, go to the web site: http://servicetoamericamedals.org/SAM/recipient/profiles/sem11_gahl.shtml



Dr. Gahl with Lara Logan from CBS' 60 Minutes. Dr. Gahl and the Undiagnosed Diseases Program will be featured on the program sometime in 2012.

CRN Announces New Members to Scientific Review Board and Medical Advisory Committee

By Colleen Hammond



Jackson Lang

Cystinosis Research Network delighted to announce the new additions to Cystinosis Research Network's (CRN) Scientific Review Board and Medical Advisory Board. Their depth of expertise in cystinosis research and clinical experience is a great benefit to the cystinosis community. We extend to them our deepest gratitude for their service.

New members of the Scientific Review Board

Robert Kleta, MD, PhD, FASN, FACMG

Professor of Nephrology and Physiology
Potter Chair of Nephrology
University College London
Royal Free Hospital
London, England

Roslyn B. Mannon, M.D.

Professor of Medicine, Division of Nephrology
Professor of Surgery, Division of Transplantation
Director of Research, Alabama Transplant Center
Birmingham, Alabama



New MAC Member Dr. Larry Greenbaum with The Smith Family at the 2011 CRN Family Conference

New members of the Medical Advisory Committee

Paul Goodyer, MD

Montreal Children's Hospital
2300 Tupper Street
Montreal, Quebec, Canada

Laurence Greenbaum, MD, PhD

Marcus Professor of Pediatric Nephrology
Emory University and Children's Healthcare of Atlanta Atlanta, Georgia

Richard H. Simon, MD

Professor and Associate Chair for Faculty Affairs
Department of Internal Medicine
Pulmonary and Critical Care Medicine Division
University of Michigan Medical School
Ann Arbor, Michigan

CRN Research Update

By Elva Smith-Vice President, Research

CRN continues to direct its focus, dedication and perseverance to the goal of securing a promising future for the cystinosis community through the support and funding of research grant studies that offer that hope. We at CRN are firmly committed to the discovery of improved treatments and ultimately a cure for cystinosis. Towards that end, CRN currently has a total research commitment of over \$800,000.00. CRN has funded nearly \$3 million in research grants and fellowships. All research grants, progress reports, funding, and final reports are provided to the cystinosis community via our web site www.cystinosis.org.



Bree and Jake Forrester

Recently completed Research Grants:

Dr. Paul Goodyer, Montreal Children's Hospital, Montreal, Canada, and **Dr. Rick Kaskel**, Children's Hospital at Montefiore, Bronx, NY. "Proposal for a North American Cystinosis Research Platform". One year grant of \$36,000.00 awarded for development.

Jess Thoene, MD, Univ. of Michigan, Ann Arbor, "Tissue Repository for Cystinosis" Grant awarded: Start date: Nov. 2006 Total award: \$26,206.00. Final report rec'd 15 Sept 2010.

Viki Kalatzis, PhD, Eric J. Kremer, PhD, Institut Genetique Moleculaire de Montpellier, France, "Gene transfer studies for cystinosis" Grant awarded 29 Aug 2007, 2 yrs. Total award: Euros 75,900.00, approx. US\$104,619.48. Final report received 25 November 2009.

Jess Thoene, M.D., University of Michigan, Ann Arbor, MI, "Feasibility of Cystinosis Replacement Therapy in Cystinosis", Grant awarded 9-25-09 for two years (9-1-09 to 8-31-11) Total Award: \$165,732. Final report received August 31, 2011, Final payment of \$55,247.00.

CRN Research Grants in Progress as of July, 2011:

Rosaleen Anderson, PhD, Sunderland Pharmacy School, University of Sunderland, "Proteomic investigation of cystinotic cells and the effects of cysteamine treatment". Grant awarded: 23 November 2010 for a term of 2 years in the amount of GBP 80,926.00 or \$130,366 based on the exchange rate for 15 November 2010.

Elena Levtchenko, MD, PhD Univ. Medical Center Nijmegen, The Netherlands, "Study of ATP metabolism in human cystinotic proximal tubular cells and in humans with cystinosis in vivo" Grant awarded: 11 September 2006. Total award: \$68,090.00 Balance remaining on grant: \$17,022.50 A "Final report" was received on 5 February 2009. However, Dr. Levtchenko has indicated that the "In vivo" part of this project is not finished yet due to the technical difficulties with the MRS apparatus, but we have good hope that the problem will be solved and this part of the study will be finished in 2009".

Catherine Tuleu, PhD, Ken Nischal, Olufemi Rabi, Rajnish Sekhri, Wm Van't Hoff, Univ. of London, School of Pharmacy; "Development of cysteamine in situ gelling system for the topical treatment of corneal crystals in cystinosis", Grant awarded January 2007 for 3 years. Total award: 103,000.00 pounds or approximately \$203,500.00. Balance remaining on grant approximately \$71,703.09.

Leticia Belmont, M.D., Unidad de Genetica de la Nutrician, Instituto Nacional de Pediatría, Mexico. "Determination of Intraleucocitary Cystine by High Performance Liquid Chromatography (HPLC) in Patients with Cystinosis". Grant Awarded: 5 November 2008 Total award: \$31,972.00; Balance remaining on grant: \$15,986.00. Presentation, citing CRN funding, was given at the IPNA meeting in NYC, 31 August 2010. Progress report rec'd Sept. 2010

Henk J. Blom, M.D., M.M.C. Wamelink, and E. Levtchenko, VU University Medical Center, Amsterdam, The Netherlands: "Newborn Screening of Cystinosis". Grant Awarded: 5 November 2008; Total Award: 45,000 euros (approximately \$67,500.00); Balance remaining on grant: 33,750 euros

Francisco Emma, M.D. and Anna Taranta, Ph.D., Bambino Gesù Children's Hospital and Research Institute, Rome, Italy: "Functional Characterization of Cystinosis-LKG". Grant Awarded: 5 November 2008; Total Award: \$114,480.00; Balance remaining on grant: \$21,465.00

CRN Research Update (Cont.)

Ewa Elenberg, MD, Texas Children's Hospital, Houston, TX "Quality of Life in Cystinosis Patients", Grant awarded 9-25-09 for 1 year. Total award: \$21,000.00 Balance remaining on grant: \$7,875.00.

CRN Research Grants Funded 2011:

CRN's Research Committee oversees the research interests of the organization and is responsible for the strategic direction in all areas of research. An official "Call for Proposals" is issued once a year and researchers are encouraged to submit their requests for funding for grants that offer the promise of achieving CRN's goals of improved treatment and a cure for cystinosis. All proposals received are reviewed by CRN's Scientific Review Board and funds are granted based on the recommendation of the Board. The 2011 grant proposal requests were reviewed by the Scientific Review Board this past July, 2011 in conjunction with CRN's Family Conference, and we are proud to announce that the following new grants have been awarded:

Maya Doyle, LCSW, Children's Hospital at Montefiore, Division of Pediatric Nephrology, New York University, "Cystinosis in Emerging Adulthood". One time grant of \$17,200.00 awarded July 2011; payment processed 17 August 2011. Study and payment complete.

Jess G. Thoene, M.D, Director, Biochemical Genetics Laboratory, Active Professor Emeritus of Pediatrics, University of Michigan, "Continuation of Feasibility of Cystinosis Replacement Therapy in Cystinosis". Grant awarded 24 July 2011 for an 18 month period for \$202,500.00. Initial payment processed 17 August 2011.

Martine Besouw, MD, Elena Levtschenko, MD, PhD; Lambertus vanden Heuvel, PhD, Univ. Medical Center Nijmegen, The Netherlands, "Study of skin changes in cystinosis patients under cysteamine therapy". Grant awarded October 2011 for a 12 month period for \$90,400.00 Co-funded by Cystinosis Foundation Ireland.



Adult men with cystinosis at the 2011 CRN Family Conference

Participants Needed for "Emerging Adulthood in Cystinosis" Ongoing Study

Participants are needed for individual interviews about the experience of growing up with and living with cystinosis as a young adult. Young adults (18-30) with cystinosis as well as parents of young adults (18-30) with cystinosis are encouraged to participate.

A researcher will come to your home or a location close to your home for the interview. Interviews last 60-90 minutes. Parents may participate in interviews only if their adult child is also a participant in the study. Young adults must agree to their parent being interviewed.

This study has received IRB approval from NYU and grant support from Cystinosis Research Network. Find out more about the study and register at <https://www.surveymonkey.com/s/yacys> or email Maya Doyle, LCSW, ABD at mhd2002@nyu.edu

Raptor Delayed Release Cysteamine Update



This past July, Raptor announced positive results from its Phase 3 clinical trial of RP103 for the potential treatment of cystinosis.

RP103 is Raptor’s proprietary, twice-daily, delayed-release formulation of cysteamine

bitartrate. The clinical trial was designed as an outpatient study of the pharmacodynamics, pharmacokinetics, safety and tolerability, of RP103 compared to Cystagon® in cystinosis patients. The clinical trial was conducted at eight clinical research centers in the US and Europe. RP103 met the primary endpoint of non-inferiority compared to Cystagon®, based on white blood cell (“WBC”) cystine levels. **WBC cystine was the sole primary endpoint in the clinical trial.** Raptor also reported that there were no unexpected safety concerns experienced by patients in the trial attributable to RP103. Forty out of 41 patients who completed the phase 3 clinical trial, elected to enroll in a long-term extension study in which they remain on the RP103 treatment.

Although publication of the clinical trial results is still pending, Raptor has presented the high-level results at the Dutch Cystinosis Foundation conference in September, in Voorthuizen (Netherlands), and the UK Cystinosis Foundation conference in Manchester (UK). Also, Dr. Elena Levtchenko, M.D., of University Hospital Leuven (Belgium), presented results from the study at the 44th Annual Scientific Meeting of the European Society for Paediatric Nephrology, in Dubrovnic (Croatia). Dr. Patrice Rioux, M.D, Ph.D., of Raptor is going to present a summary of RP103 development at the 9th Latin American Congress of Pediatric Nephrology (ALANEPE) in Sao Paulo (Brazil) on October 29, 2011, and Dr. Craig Langman, head of kidney diseases at Feinberg School of **Medicine, Northwestern University, and principal investigator in Raptor’s phase 3 clinical trial,** is scheduled to present the results in a poster presentation at the American Society of Nephrology Kidney Week, in Philadelphia, on November 12, 2011.

In addition to its Phase 3 clinical trial of RP103 in cystinosis patients, Raptor conducted a bioequivalence clinical trial of RP103 in healthy volunteers, which demonstrated that the drug may be administered by sprinkling the contents of the capsule onto applesauce. Based on these findings, Raptor plans to expand enrollment in the RP103 extension study, to include small children who are not able to swallow whole capsules. Raptor also plans to enroll in the extension study, transplanted patients who were not eligible for the phase 3 clinical trial. The company expects its clinical sites to begin enrolling these additional patients before the end of this year.

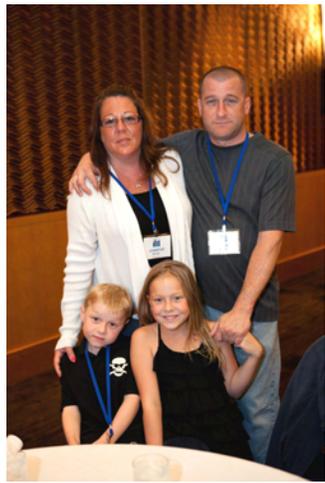
Meanwhile, Raptor will be meeting with US Food and Drug Administration (FDA) in late October, and with European Medicines Agency (EMA) in early November, to discuss its plans to submit applications for marketing approval of RP103 to treat cystinosis. Pending positive outcomes of those meetings, the company expects to file for marketing approval in the US and Europe, in the first half of 2012. Depending on review timelines by FDA and EMA, Raptor hopes for marketing approval and commercial launch of RP103, before the end of 2012.



The Stanford Family



Joe and Caitlin Jordan



The Binder Family

CRN with Sigma Tau Q&A



Kenadee (with cystinosis), Jan, and Taylee Julian

Can you give us a brief history of how Sigma-Tau became involved with cystinosis?

In 1995, The National Organization for Rare Diseases (NORD) approached Sigma-Tau Pharmaceuticals with a special request to help the National Institutes of Health (NIH) secure a GMP (good manufacturing practice) active pharmaceutical ingredient for cysteamine eye drops. Over time, the collaboration grew and Sigma Tau became involved in regulatory planning and manufacturing of the eye drops. Also Sigma Tau began to build partnerships with the cystinosis patient groups. In 2004, Sigma-Tau assumed regulatory responsibilities and scientific leadership for filing to FDA a new drug application (NDA) for the cysteamine eye drops.

Why wasn't Cystaran® FDA approved in September 2011 as they were thought to be?

There were technical issues surrounding the manufacturing of the product through our third party manufacturer that delayed the decision. We were issued a complete response letter in September 2010 that afforded us and our third party manufacturer the opportunity to rectify the technical issues.

We understand there were some manufacturing challenges for Cystaran®; can you let us know where you stand with those issues?

The FDA was concerned with the overall GMP compliance of the facilities involved in the production of the drug substance and the drug product. FDA did not question the manufacturing processes of Cystaran, but the overall GMP (good manufacturing practice) compliance of the facilities involved in the production of the drug substance and drug product. We are working with the facilities to address these issues and to resolve them in the near future.



Jenni and Josie Sexstone

When will Cystaran® be FDA approved?

Once the GMP compliance issues are resolved, FDA will review the CMC (chemistry, manufacturing and controls) updated information. If the Agency is satisfied, the product could be approved.

Once resolved, how long will it take to actually get Cystaran® to the patients?

We expect three-to-six months.

Where will patients obtain Cystaran®?

Once Cystaran® is approved; availability will be managed through a specialty pharmacy which will secure insurance coverage and then ship the medicine directly to patients.

In the interim, should patients continue to get their eyes drops from the NIH?

NIH has agreed to continue to make the product available under a specific clinical trial protocol. Patients need to be enrolled in this NIH protocol to receive the eye drops.

CRN with Sigma Tau Q&A (cont.)

Some people currently have their eye drops compounded at a compounding pharmacy; will they be able to obtain their compounded eye drops from those pharmacies once Cystaran® is approved?

The FDA-approved product would only be available through a specialty pharmacy.

Currently, patients are instructed to administer 1 drop in each eye every waking hour. Will that still be the recommended dosing for Cystaran®?

Yes, that was the recommended dosage standard during the supportive clinical trials.

Will Cystaran® still need to be stored frozen/ refrigerated?

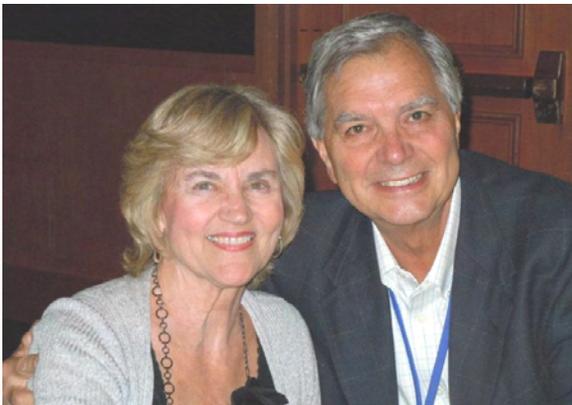
The product easily oxidizes to cystamine if not stored frozen or kept refrigerated after opening. Unopened Cystaran® bottle must be kept in the freezer for storage. The bottle must be thawed for 24 hours before use. After opening, the bottle can be kept refrigerated (36°F) or at room temperature (77°F) for one week.

How much will Cystaran® cost? Will it be covered by insurance and what if a patient has no insurance or cannot afford Cystaran®?

We have looked into many potential outcomes for insurance coverage, and while we are confident that coverage for most patients will not be an issue, Sigma-Tau has established a very comprehensive patient assistance program for all of our medicines, some include 100% copay assistance. If Cystaran® is approved; it will also be included in this program. We have not determined a price point for Cystaran® at this time.

The relationship with Sigma-Tau and the cystinosis community has been a long-standing one. Will Sigma-Tau continue to be involved in the cystinosis community once Cystaran® has been approved?

Sigma-Tau plans to continue our partnerships with the cystinosis community for many years to come. Once we have a final decision on Cystaran® we hope to focus efforts toward the development of an improved product using a slow-release technology with the intent to reduce the frequency of eye drops administrations.



Brenda and Tom Melang



The Binger and Stone Families



Sookie Boyer



Jose Morales, Francois Coupey, Dan Julian, and Beatrice Coupey

CRN Financial Update

By Jeff Larimore-Treasurer



Katie, Jeff, and Sarah Larimore

For the 9 months ended September 30, 2011

We are pleased to present the financial results of the dynamic activities of the Cystinosis Research Network through September 30, 2011. With the focused and concerted efforts of the CRN membership, many great events have been held and outreach awareness has been extended around the globe.

For the nine months ended September 30, 2011, total income secured of \$299,000 was approximately 40% greater than the same period in 2010. The increase was derived from improved fundraising efforts from dinner dances and sporting events plus grants received to support the Family Conference. Total donations received from public organizations such as United Way, Network for Good and Facebook were approximately \$39,000 which was consistent with 2010.

Administration, registration and travel costs associated with domestic and international education awareness along with the 2011 Family Conference amounted to approximately \$152,000 through September 2011. In 2010, CRN provided a \$50,000 sponsorship for the International Pediatric Nephrology Association conference that was held in New York City. General operating expenses for CRN through September 2011 of \$29,000 have decreased approximately \$60,000 as compared to the ninth months ended September 2010.

In addition to Education and Awareness, a primary objective of the Cystinosis Research Network is to provide funding for research grants. Grant payments of \$370,000 have been made through September 2011. The institutions that have received support from CRN in 2011 include:

- University of Sunderland
- University of Michigan
- National Human Genome Institute
- University of London
- New York University
- Baylor University

For the period ending September 30, 2010, approximately \$125,000 of grant commitments was paid. Throughout the newsletter will be more detailed information regarding each of the research programs in progress.

Continued on page 15

CRN Financial Update (cont.)

A challenge that lies ahead for the Cystinosis Research Network is the funding of the research grant commitments that have not yet been progress billed to the organization. Over the next few years, approximately \$394,000 of research grant commitments will need to be paid. In order for CRN to help those researchers and physicians who **are focused on helping our children, adults and families, CRN’s membership must be equally committed to sustaining its advocacy group through fundraising and donations.**

A comparable financial recap for Cystinosis Research Network for the nine months ended September 30, 2011 and 2010 is:

	<u>2010</u>	<u>2011</u>
Total Income	\$299,000	\$214,000
Total Operating Expenses	<u>(577,000)</u>	<u>(278,000)</u>
Net Operating Income (Loss)	<u>(\$278,000)</u>	<u>(\$64,000)</u>
Cash on Hand – January 1	\$477,299	\$461,841
Net Change in Cash	<u>(306,156)</u>	<u>(83,310)</u>
Cash on Hand – September 30	<u>\$171,143</u>	<u>\$378,531</u>

As presented above, the successful activities of the Cystinosis Research Network has made a dent in its operating cash during 2011. One of the goals discussed by the Executive Committee and the Board of Directors in San Francisco was to put together and implement strategies that will help minimize the large fluctuations in operating cash during the years that the Family Conference is held. A continuing goal of the organization is to reach \$500,000 in total income. Through September 2011, three events have cumulatively raised over \$150,000. What if those **events weren’t held??**

A quote from Victor Kiam, founder of Remington shavers and former owner of the New England Patriots, was **“Procrastination is Opportunity’s natural assassin”.** Let’s assure ourselves that we don’t wait to be prepared financially to miss that **“end of Cystinosis” opportunity.**



Rekindling friendships at the 2011 CRN Family Conference:

Jakob Ellerbrock, Josie Sexstone, Jordan Sexstone, Sarah Larimore, and Kylie Ellerbrock

CRN Education and Awareness Update

Upcoming Family and Medical Events

November 8-13, 2011

ASN Renal Week
Philadelphia, PA

November 15-16, 2011

NORD Board of Directors Meeting
Washington, DC

December 2, 2011

NORD Regional Meeting
New York, NY

February 29, 2012

Rare Disease Day

April 28-May 1, 2012

2012 PAS/ASPN Annual Meeting
Boston, MA

June 20-24, 2012

Genetic Alliance Conference
Bethesda, MD

Oct. 30-Nov. 4, 2012

2012 ASN Renal Week
San Diego, CA

By Paula Shal Vice President- Education and Awareness

CRN is fortunate to have a core group of board members who are willing to donate their time to travel to medical meetings and events in order to bring cystinosis to the forefront of the medical community.

I just wanted to take a moment to recognize these individuals who literally **"pound the pavement" to get the word** out about cystinosis—Karen Gledhill, Christy Greeley, Colleen Hammond Marybeth Krummenacker, Frankie McGinnis, Jose Morales, Elva Smith, and Pam Woodward are our road warriors. Sometimes they go with very little notice, and sometimes they even donate their own travel expenses. Their photos are in the newsletter often because they

have dedicated a large part of their lives to finding better treatments and ultimately a cure for a small, unknown, rare disorder with no government funding. A key to making **progress toward CRN's mission and vision is** networking with organizations and individuals. Help is out there, and this group knows how to sniff it out.

I am also excited to announce that Katie Ahnen, Mikaela Gard, and Weston Tschannen are all recipients of CRN academic scholarships for individuals with cystinosis! These three inspirational young people will be fully featured in our next newsletter.

CRN hopes you enjoy this edition of The Cystinosis Advocate and as always, if you have any newsletter suggestions, please feel free to e-mail me at pschal@cystinosis.org.

CRN Sponsors Cystinosis Symposium at the 2011 Latin American Pediatric Nephrology Association Meeting in Sao Paulo, Brazil

The Cystinosis Research Network co-sponsored the 2011 Latin American Pediatric Nephrology Association meeting (ALANEPE 2011) in Sao Paulo, Brazil on October 26. The meeting was chaired by Dr. Vera Koch from Brazil and Dr. Leticia Belmont from Mexico, both of whom are involved in treating patients with cystinosis and cystinosis research. Both women are passionate advocates for their patients, and especially showed their dedication to the cystinosis community by organizing a half day session on the disease.

Physicians and students from all across Latin America attended the morning symposium, which was moderated by Dr. Frederick Kaskel and included speakers who not only educated the attendees on the diagnosis and treatment of cystinosis, but also offered updates from different areas of Latin America and gave the patient advocacy perspective in the U.S.

CRN was proud to not only underwrite a portion of the meeting, but to also sponsor Dr. Galina Nesterova, CRN NIH Clinical Fellow, and Dr. Doris Trauner as speakers. Christy Greeley and Jose Morales attended as representatives of CRN, where they were able to not only give the audience the patient and family perspective from the U.S., but also to communicate the vital importance of patient advocacy groups supporting the wide array of needs of their patient population. Funding research is key, but on a daily basis, education and awareness and family support activities are often the most valuable services we provide. We will continue to assist with the development of patient advocacy groups in Brazil and the rest of Latin America as a demonstration of CRN's dedication to supporting patients in the U.S. and throughout the world.



Victor and Maryan Gomez

CRN Sponsors Cystinosis Symposium at the 2011 Latin American Pediatric Nephrology Association Meeting in Sao Paulo, Brazil (Cont.)



Agenda/Cystinosis Symposium

Introduction	FREDERICK J. KASKEL (CHILDREN'S HOSPITAL AT MONTEFIORE, ALBERT EINSTEIN COLLEGE OF MEDICINE, NEW YORK)
Early presentation and pathophysiology	PAUL GOODYER (MONTRÉAL CHILDREN'S HOSPITAL, CANADA)
Late manifestations and outcomes	GALINA NESTEROVA (OFFICE OF RARE DISEASES OF NATIONAL INSTITUTES OF HEALTH – NIH, USA)
Developmental and neurocognitive issues	DORIS TRAUNER (UNIVERSITY OF CALIFORNIA, HOSPITAL SAN DIEGO, CA USA)
Stem cell and gene therapy for cystinosis	STEPHANIE CHERQUI (SCRIPPS RESEARCH INSTITUTE, LAJOLLA, CA USA)
Overview of Latin America diagnostic and therapeutic possibilities	LETICIA BELMONT (INSTITUTO NACIONAL DE PEDIATRIA), NATALIA MEJIA GAVIRIA (UNIVERSIDAD DE OVIEDO), VERA KOCH (PROFESSORA LIVRE DOCENTE, FMUSP, INTITUTO DA CRIANÇA)
Transition of care from pediatrics to adult	FREDERICK J. KASKEL (CHILDREN'S HOSPITAL AT MONTEFIORE, ALBERT EINSTEIN COLLEGE OF MEDICINE, NEW YORK)
The Cystinosis Research Network	JOSE MORALES AND CHRISTY GREELEY
The Cystinosis Research Foundation	NANCY STACK
The Cystinosis Foundation	VALERIE HOTZ
CCIR – CCIR in Portuguese and Spanish	VERA KOCH (PROFESSORA LIVRE DOCENTE, FMUSP, INTITUTO DA CRIANÇA)
Cystinosis Mexico	VICTOR GOMEZ
Open Forum —Panel of government representatives (Secretary of Health and Secretary of the rights of patients with special needs), health professionals, affected children and their families, and patient advocacy groups	CARMELA MAGGIUZZI GRINDLER (COORDENADORA ESTADUAL DO PROGRAMA NACIONAL DE TRIAGEM NEONATAL DE SÃO PAULO)



Jose Morales, Dr. Galina Nestrova, Dr. Frederick Kaskel, Dr. Vera Koch, Dr. Paul Goodyer, Christy Greeley, and Dr. Leticia Belmont at the 2011 Latin American Pediatric Nephrology Association Meeting



Christy Greeley, Dr. Frederick Kaskel, and Dr. Galina Nestrova in Sao Paulo, Brazil

Report from the National Organization for Rare Disorders



By **Marybeth Krummenacker**



Marybeth Krummenacker with Dr. Jess Thoene at the 2011 CRN Family Conference

The National Organization for Rare Disorders (NORD) is acknowledged globally as the leading nonprofit organization representing the nearly 30 million Americans with rare disease. It is truly an honor and a privilege to represent the Cystinosis Research Network and the entire cystinosis community at important monthly meetings both in person and in telephone conference calls. I have been asked to serve on the membership committee in addition to being a member of the advocacy committee. NORD is a definitive presence in Washington D.C. and highly respected by industry and government officials. When it comes to advocating for the rare **disease community.....very often policy makers turn to NORD for advice, guidance and opinion.** With the advances in science and technology, it is hard to keep up the pace, but NORD and its staff are visible on the front lines of change.

NORD's annual Corporate Council meeting and Gala was held in the Mellon Auditorium in Washington, D.C. in May, 2011. Christy Greeley, Jose Morales, Maya Doyle, and myself attended this event to celebrate the accomplishments many in the rare disease community and to honor those who continue to go above and beyond.

On September 16, Christy Greeley and Jose Morales spoke at the NORD Regional Meeting in Chicago. They highlighted the issue of organizational development as it relates to development and overall governance of a non-profit organization. They brought over 12 years of experience to a discussion about everything from finances to forming an advocacy group board. Again, it was a unique experience for CRN to be one of the key organizations represented at this first regional meeting. The next meeting will be held in New York on December 2, and CRN will again have representation in attendance.

On October 11, 12 and 13, I attended the first "Rare Disease Summit" in Washington D.C., where for the first time leaders in the communities of the pharmaceutical industry, government, and advocacy came together. These groups brainstormed and learned about issues in the rare disease community with a common goal of trying to understand the gaps that exist that may prevent advancement. This was an opportunity to network with some leading decision makers and to hear about the advances in science and research and how it relates to rare diseases. Those who live with rare diseases like cystinosis every day were able to share their own stories as well as the stories of their community with people like Dr. Francis Collins, Director of the National Institute of Health. It was also an opportunity to try to understand the process of qualifying for SSD or SSI and the Compassionate Allowance program by speaking directly to Commissioner Michael Astrue of Social Security.

We live in a fast-paced times with discoveries happening every single day, and there is much to be learned by sharing stories and real life experiences with people who not only listen but understand the realities for those living with rare diseases. The diseases may be different, but there are over 7,000 rare diseases. The challenges, questions, answers to living with **these diseases are all similar and need to be told. Please don't be afraid to tell your stories.** It does have impact, and people do listen.

Kathy Mandrell Attends Genetic Alliance Annual Conference: 25 Years of Innovation

By Kathy Mandrell

I was thrilled this summer to be asked to represent CRN at the Genetics Alliance Conference in Bethesda, Maryland. It just so happened that I had a work conference to attend that same week in Bethesda just four miles down the road, so I was able to combine the trips and the conferences only overlapped by a smidge. Let me tell you, I learned a lot that week!

The conference celebrated 25 years of innovation in the genetics and health communities. Thursday evening, we were treated to a series of short films called Gene Screen. The films were informational, inspirational, and frequently emotional. You can see a few of this year's and previous years' short films at www.geneticalliance.org/genescreen2011. On Friday, I attended a full-day symposium on Genetics Services. Collaboration was the theme and among several presenters, I was very impressed with the HALI Project (www.thehaliproject.org). The Thompson family's mission and purpose is this project which brings together parents of special needs children, primary care providers and educational institutions as a team to serve families of children with special needs. They have several programs which provide education and counseling to new parents, day camps for special need children and their siblings and even marriage retreats for the stressed-out parents.

Surprisingly, the Policy Outcomes: Incentivizing Communication and Support topic gave me the most to think about. Now I say "surprisingly" for me, as I'm not a very politically-minded or outspoken person. TJ Sutcliffe, from www.thearc.org made this presentation. She emphasized the need to teach people how to communicate with their governments in order to affect change. What legislators need most is information to use when they make their arguments: family stories and facts about life with disease and/or disability. Besides legislative change, the general public also needs to know how disabilities affect their friends, colleagues and neighbors. If they don't know about it, they can't do anything to help. Ms. Sutcliffe said, "Every person you meet every day is an opportunity to educate. You never know what effect they can have".

As always, networking is one of the most important functions of any conference. I had lunch with Lara Mangravite of Sage Bionetworks in Seattle and Brian Loew, the founder of Inspire. Sage (www.sagebase.org) is a 501(c)(3) non-profit biomedical research organization which acts as a link for academic and industry researchers to apply genetics research to improve disease therapies. Inspire (www.inspire.com) is a social networking site where people dealing with serious disease can share with each other. It also allows pharmaceutical companies to inform this highly engaged population about clinical trials that are available to them, hopefully boosting enrollment. Finally, there was a display table set up outside one of the seminar rooms that I just had to visit. It was titled simply "Knitting". There, you could borrow a set of needles or a crochet hook and some yarn and spend your seminar creating a work of art. I made a spiral scarf and Denise Shereff from the University of South Florida made several cute flowers for hats and bags.

Thanks to the Cystinosis Research Network for giving me the opportunity to attend this conference. Be sure to check out the Genetic Alliance website if you want to see more of what they do at www.geneticalliance.org.

Save the Date!



Rare Disease Day
February 29, 2012



Mary Peckiconis, GA staff; Hannah Dolins, Maple Sugar Urine Disease; Kate Halbruner, GA staff; and Denise Shereff, USF at the knitting table.

Cystinosis Foundation Ireland Hosts Symposium, Fundraises, Reaches Milestone

By Sue Maguire



Lady Mayor of Galway City and County poses with a group of cyclists prior to the coast to coast cycle

Cystinosis Foundation Ireland is pleased to announce that our inaugural Research Symposium was held on October 13 in the prestigious Conway Institute, University College Dublin.

Uniquely, all of the talks at the symposium were presented in **lay persons' terms. We specified this because we issued an open invitation to all our members and supporters. We wanted them to be able to learn more about the research they are helping to fund in terms everyone can understand.**

Seven speakers were invited from University College Dublin, University College Cork, Robert Gordon University, Aberdeen and Stanford University California. All present had their research funded by Cystinosis Foundation Ireland and many have also received funds through a Joint Funding Scheme with the Health Research Board (HRB).

This meeting came at a very significant time for us as we also announced that since our inception in 2003 we have now given 1 million Euros toward cystinosis research! We are extremely proud of this milestone as we feel it is a wonderful achievement for our small group who have always remained focused on our aim of improving current treatments and ultimately finding a cure for the disease.

We have had a busy year on the fundraising front with lots of fun events and many new friends made throughout the year. We hosted our annual events, such as our Golf Classic and Women's Mini Marathon, and this year we added a coast to coast cycle, tandem sky-diving, and busking (street performing)! Our annual fundraising target is 100,000 Euros, and we are pleased to say thanks to the help of all our friends and supporters, we should meet our budget once again.



CF-Ireland Executive Committee at the CF Symposium celebrate donating one million Euros toward cystinosis research since its inception

Cystinosis Foundation Ireland Symposium Agenda

Time	Presenter	Title
14:00-14:10	Anne Marie O'Dowd, Chair of Research Group, Cystinosis Foundation Ireland	Cystinosis Foundation Ireland Welcome
14:10-14:40	Patrick Harrison, Dept. Physiology, University College Cork	100 years of cystinosis – one drug, no cure, and a lot of unanswered questions
14:40-15:00	Katrin Kaschig, Dept. Physiology, University College Cork	Cystinosis Gene Repair I: How to check, if the function of the CTNS gene has been restored
15:00-15:20	Ciaran Lee, Dept. Physiology, University College Cork	Cystinosis Gene Repair II: Generation of new tools to excise the defect CTNS gene
15:20-16:00	Poster Session and Refreshments	
16:00-16:20	Rodolfo Sumayao, University College Dublin	How can laboratory cell line studies help us to understand cellular dysfunction in Fanconi syndrome in cystinosis?
16:20-16:40	Bernadette McEvoy, University College Dublin	Cystinosis affects pancreatic beta cells, the cells that make insulin in the body
16:40-17:00	Graeme Kay, Robert Gordon University, Aberdeen	<i>Unfortunately Dr. Kay has had to withdraw for family reasons</i>
17:00-17:20	Poonam Sansanwal, Stanford University	Identification and characterization of novel cellular injury molecules and pathways in cystinosis
17:20-17:30	Cystinosis Foundation Ireland	Chairman Summary
17:30-18:00	Poster Session and Discussions	

A group of cyclists celebrate finishing the race from the West coast of Ireland to the East coast



CRN Family Support Update

By Pam Woodward
Vice President-Family Support

I want to start by giving a big "Thank You" to Cheri Friend who served as Vice President of Family Support for the last two years. She did a wonderful job and her services were greatly appreciated. The Family Conference in San Francisco on July 14--16, 2011, was a huge success and she did another fantastic job with the registration and conference information binders. We were able to give 10 scholarships for the Conference thanks to C.H. Robison Salt Lake Branch earmarking some of the proceeds from the Annual Golf Tournament that they hold for Cystinosis Research Network. The 2013 Family Conference is already shaping up nicely and will be held in the Washington D. C area. Start planning now so that you can be in attendance. We are hoping to be able to again offer conference scholarships to those wishing to attend to help with the cost of the lodging. Since 2012 will be an off year for the conference it is the perfect time for regional gatherings. If anyone is interesting in hosting one, please contact me at pwoodward@cystinosis.org.

The Adult Care Excellence Initiative that is under the Family Support Committee completed the Pediatric to Adult Care Transition Guide and continues to strive toward meeting its goals. The initiative is divided into 3 subgroups: 1) Clinical Practice Guidelines for Cystinosis – Pediatric through Adulthood, 2) Transition from Pediatric Care to Adult Care, 3) Identify Research and Treatment Needs to Improve the Quality of Life for Adults with Cystinosis. Please see below for more more information on this initiative.

I also have some extra copies left over from the conference of the book "Without a Word" by Jill Kelly wife of NFL Quarterback Jim Kelly whose son was born with a rare genetic disease. If you are interested let me know.

I Hope everyone has a wonderful Holiday Season and a Happy 2012.

CRN Adult Care Excellence (ACE) Initiative



Tahnje Woodward, Jessica Jondle, and Laura Krumenacker at the 2011 CRN Family Conference

By Colleen Hammond

The Adult Care Excellence (ACE) Initiative Committee was delighted to present the Pediatric to Adult Care Transition Guide at the CRN Conference in San Francisco. The Guide is available on the CRN website at: <http://www.cystinosis.org/what-is-cystinosis/resources/transitioning-guide>. The Guide continues to be a work in progress. We would greatly appreciate your feedback regarding other issues you would like included in the Guide.

We continue to work to provide resources for the cystinosis community. Our goals for this year include:

- Developing a Booklet of the Standards of Care for Children and Adults with Cystinosis. We are working with Dr. Gahl and Dr. Nesterova at the NIH. The document will be available on the CRN website for families, adults, and physicians to download.
- Guide for Parents of Children with Cystinosis. This Guide will be similar to the Transition Guide, but will be directed to parents of children with cystinosis from birth through age 14.
- The ACE Committee will attend Transplant Physician Medical Meeting to raise cystinosis awareness.

Gabe Stephenson Inspires: "Keep on Running!"



In September 2010, Gabe Stephenson competed in a bench pressing competition. He set the Kentucky state record for 14 year old males in his weight division. He weighed in at 105 pounds and successfully benched 115 pounds! His 2011 goal is 125 pounds.

On May 7, 2011, Gabe completed his first 5K run in 26:35 minutes. Next year, he plans to break this personal record. In training, he is running 4 miles on a treadmill in less than 35 minutes and plans to be at 5 miles in 35 minutes by spring.



Gabe Stephenson

Gabe says it is important that he continues to meet these physical goals to encourage other people with cystinosis to not let the disease hold them back...Keep on running!

William Tschannen Receives 2011 CRN Sierra Woodward Sibling Scholarship



William Tschannen

CRN awarded its annual sibling scholarship to William Tschannen, brother of Weston Tschannen, who has cystinosis. William is currently in the honors college at Missouri State University and studying Cellular and Molecular Biology. His goal is to be admitted into medical school and become a doctor.

"I want to be a doctor so that I can help people," Tschannen said, "Just as other doctors have helped my family for so long. I don't know what life would be like without my twin brother and Cystinosis, but I am thankful for such a great family. We have all had our bad days, but we have stuck through it together and have always been there for each other. "

Jessica Jondle Writes Book About Growing up with Cystinosis



Jessica Jondle was diagnosed with cystinosis at the age of 22 months and is now approaching the age of 30. She has written a book called *Roller Skating with Rickets* (and other paradoxes of life with genetic disease).

Jessica graduated from UC-Berkeley and is working on her Master's degree in Education. She teaches middle school history and is the upper school Academic Dean at an accelerated K-8 school.

Jessica is anxious for her book to be available to the public. You can find out more about the book and reserve a copy now at www.rollerskatingwithrickets.com.

Midwest Cystinosis Gathering Carries on the Tradition

By Doretta Hoffman

The 22nd annual Midwest Cystinosis gathering was held on July 29 – 31 in Des Moines, IA. We had a great time again this year with 30 people attending and a special guest, Maya Doyle.

On Friday evening after we all went together to eat, and then we visited around the pool area while a few did some swimming. This was a great time to “catch up” on what had happened in our families during the past year.

Saturday morning some of the families decided to attend the local farmer’s market. All of the people that attended had a great time and some even tried the Dutch letters (a type of almond cookie) and thought they were a good purchase. Later in the morning we all met with Maya and talked about the transitioning project emerging into adulthood study. Maya also interviewed the cystinosis young adults that were in attendance at the gathering.

After lunch the group headed off to the Des Moines Science Center for the afternoon. Some attended the IMAX movie of Harry Potter and others went to the Human Body Worlds traveling exhibit and learned all about the muscles and organs of our bodies. Saturday afternoon we watched a video of the national CRN conference in San Francisco for the families that were unable to attend.

Saturday evening we all went to the Iowa Machine Shed restaurant which is a local attraction for dinner. The staff at the Machine Shed sang happy birthday to Nick Wagner for his 28th birthday. Some of the group went on a hay ride thru the living history farm area before heading back to the motel for more visiting.

Sunday morning we said our good byes and departed. Brad and Gretchen Thurman have **volunteered to host next year’s gathering in the Indianapolis, IN area.** Hopefully, we will be able to have some new families join us. Everyone is welcome!

One final note- the Midwest Cystinosis group missed one of our members, Darlis Croce, this year. Darlis passed away in February. She was a wonderful wife, mother, and friend. The group presented the Croce family with a remembrance book with pictures of Darlis when she had attended our past cystinosis gatherings. Darlis was a dear friend to our group and is missed by all of us. Please keep the Croce family in your prayers as they continue to go thru life without Darlis.



Bill Sr. and Bill Jr. Croce

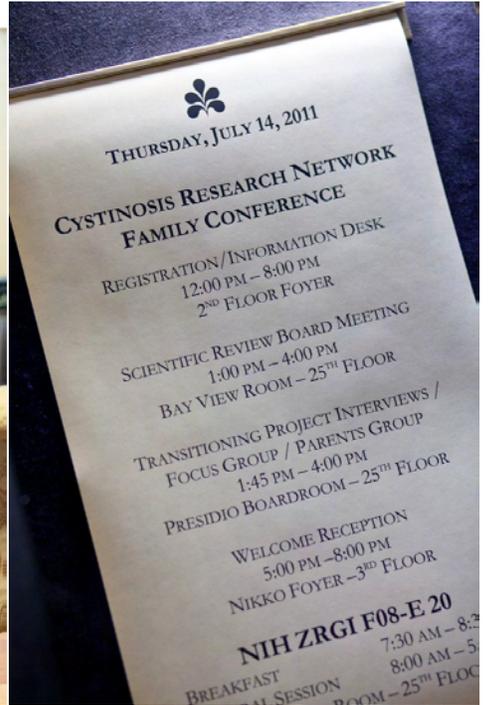


Front Row - Wyatt Thurman, David Reuter, Shawn Reuter, Mikaela Gard

Back Row - Andrew Hoffmann, Weston Tschannen, Nick Wagner

2011 CRN Family Conference Scrapbook

Registration



Welcome Reception





Family Introductions



Group picture of everyone who attended the 2011 CRN Family Conference



The Alexander Family



The Schleuder Family with Karen Kuphal from Sigma Tau and Katie Freed from SW Medical Research Foundation



The Woodward/Boyer Family



The Reed Family



The Gilje Family



The Ellerbrock Family

Speakers and Workshops



Don't forget to check out [Part 2](#) of The Cystinosis Advocate, which features great fundraisers and more conference photos!