The Cystinosis Foundation Newsletter
Volume 18, Number 1
Fall 2010

A Report on the 6th International Cystinosis Congress

By Marjolein Bos
Dutch Cystinosis Group

We attended the 6th International Cystinosis Conference in Italy with a group of 12 persons from The Netherlands . Five individuals of this group are young adults dealing with cystinosis, three of these young adults are still students. For these students we have found a sponsor to pay for their travel expenses

[See REPORT page 18]

Marjolein Bos leads group discussion.

INSIDE

Colin Daniels, William A. Gahl, MD, and Gail Daniels cross paths at Frankfurt Airport.

Our First Cystinosis Conference

At Last, A Pin On The Map For South Africa

By Gail Daniels

In Transit

Being in transit can be exhausting and especially after 20 hours of travelling. Sitting at Frankfurt airport for seven hours, pacing up and down waiting for our connecting flight, I looked up and saw a face that looked so familiar. "I have seen this face before, I know this man, I am certain it is him," I thought to myself.

I rushed back to my family and with great excitement told them, "Guess who I just saw?" Their tired faces lit up when I said "Dr. William Gahl", a name that is mentioned so many times at home and a face I had seen in many Cystinosis Foundation newsletters and related articles. I was so excited I could hardly breathe. I plucked up the courage and went straight over to him, stood in front of him and said, "Excuse me sir, are you Dr. William Gahl?" He stood up, stretched his hand out and said "Yes, I am". "Wow, what a pleasure and honour to meet you" I replied. For a few minutes we chatted and had his undivided attention. There was just one thing; he spoke through his teeth. Literally, he spoke through his teeth. He saw our confusion and explained that he had broken his jaw.

With eight months of planning behind us, bags packed and two days of travelling across our continent we eventually arrived in the Village GeTur, Lignano Sabbiadoro to experience our first International Cystinosis Conference. As we watched the delegates arrive in their masses, our feelings fluctuated from excitement and joy to being overwhelmed seeing so many strangers and foreigners yet in some weird and wonderful way there was a feeling of belonging.

Meeting our 'cyber mates'

The excitement and awesomeness of seeing a familiar face 'come to life' was so satisfying. Friends that we have been chatting with via emails and the Family Forum, were now standing right in front of us with outstretched and welcoming arms. Wow, that was pretty amazing! It was really nice meeting some 'in colour' for the first time after only seeing black and white

[See FIRST page 18]

From the President's Desk

We recently celebrated the 28th anniversary of the founding of the Cystinosis Foundation with the 6th International Cystinosis Congress and I am reminded of Dr. Thoene, Dr. Gahl and Dr. Schneider's devotion and dedication to our community, which has been extended worldwide over the past few decades. They have helped us embrace our international community and today we truly reach beyond borders .

Dr. Schneider was our inspiration, our trusted advisor and mentor from Day One. We learned the importance of connecting with other organizations from Dr. Thoene and consequently became the first members of the newly established National Organization for Rare Disorders and also the Genetic Alliance. Dr. Gahl has always been a constant source of encouragement, as he understands the importance of our need to reach out to families wherever they live in the world. We have seen tremendous strides in cystinosis research as a result of a strengthened and expanded international scientific community. Throughout the years these scientists have shared their expertise with families, as well as the international medical community at unique Cystinosis Foundation family conferences.

As a result of this collaborative spirit, cystinosis research crossed international boundaries years ago. Dr. Michel Broyer, Dr. Corinne Antignac, Dr. Stephanie Cherqui, Dr. William van't Hoff, Dr. Elena Levtchenko, Dr. Francesco Emma, Dr. Doris Trauner, Dr. Ranjan Dohil, the late Dr. Gianfranco Rozzini and others have participated in increasing the understanding of the mechanisms of cystinosis and a new generation of great scientific minds are stepping up. We embrace them enthusiastically.

As we begin the second decade of the second millennium, we salute the launch of The Cure Cystinosis International Registry (CCIR). We urge you to register if you have not already done so. With every registration, scientists will gain more information about this rarest of rare diseases. You can be an active contributor to research progress without the hassle of traveling. Privacy safeguards are in place to protect the privacy of each patient who registers. By registering now, you can personally contribute to the progress of scientific research that will lead to improved treatments and one day a cure for this rare disease. Registered individuals may gain some peace of mind knowing the experts within the cystinosis medical community have all specific details about their health (and for parents, their child's health) as contained within the registry.

Many doors have been opened by researchers and you can do your part to help open additional doors by registering with CCIR. The knock-out mouse has been developed which scientists are utilizing in advancing research and the gene responsible for cystinosis was identified in 1998. Clinical trials for a delayed release form of cysteamine requiring dosing every 12 hours are taking place now in the U.S. and Europe, raising our hope for approval by the Food and Drug Administration and the much needed extra sleep that will follow. We anticipate official FDA approval of Sigma-Tau's eye treatment soon.

In the British Virgin Islands they celebrate Old Year's Day on December 31 by reminiscing about the events that have

[See PRESIDENT page 4]

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The Cystinosis Foundation is always looking for new and interesting articles to publish in our Newsletter. There are many topics of interest to the cystinosis community from both a medical perspective and also from a personal point of view. If you are interested in having an article published in our newsletter or wish to submit a letter to the editor, please send it to: Valerie Hotz at v.hotz@att.net. We look forward to hearing from you.

The Importance of Play

By Don Hammond


Don Hammond and Shawn Reuter build castles in Lignano.

What happens when youth from greatly differing cultures come together with the common uniting factor of cystinosis? Some have cystinosis, some are siblings and others are friends. So many differences and yet each youth is still facing the same tasks of emerging into adolescence and adulthood.

During this year's conference these amazing kids and young adults talked about their lives, their hopes and dreams. They also talked about how they have been impacted by living with cystinosis. Each individual struggles to move toward independence, each trying to manage relationships, education and figuring out what their lives will become. Some of our guests at the congress are venturing off to college, living away from home for the first time and growing in their independence. It is exciting to see them seize life and storm full speed ahead.

Some of them just needed to have fun. Have you ever thought about how important it is just to laugh and play? It's a great release from the stresses of life. So one of the big things we did at the International Cystinosis Congress in Lignano was just play. We ventured to the nearby beach and facing the Adriatic Sea we built sand castles and played with colorful striped beach balls. Playing together tears down separating differences, establishes trust and in so doing becomes a foundation for building our relationships.

Hopefully the kids that spent time playing and laughing together in Italy came away with a deeper sense of well being and wellness that will serve them on their journey.

Editor's Note: For more photos of children playing at the conference, please see page 8.

Mikhail Kagan, MD to Visit Belgium to Participate in Cystinosis Clinic

By Elena Levtchenko, MD

Dr. Mikhail Kagan, pediatric nephrologist at Orenburg Regional Children's Hospital in Russia diagnosed the first patient with cystinosis in his country in 2007. Since that time Dr. Kagan has become an active member of the cystinosis community. Thanks to the invitation and financial support of the Cystinosis Foundation, Dr. Kagan visited two International Cystinosis Congresses in Dublin, Ireland in 2008 and in Lignano, Italy in 2010. Due to his efforts and those of Alexey Tsygin, MD, professor and Chairman of Pediatric Nephrology Department in Russian Academy Research Center for Children's Health in Moscow, physicians in Russia became aware of cystnosis. In the meantime, seven additional patients with cystinosis were diagnosed in the span of just two and a half years.

Russian physicians struggle for the optimal care of their patients despite the absent regulation for Cystagon importation to Russia, difficulties in obtaining supportive treatment and eye drops, and no availability of biochemical or genetic diagnostic tests for cystinosis.

In order to gain more clinical experience in treating cystinosis patients, Dr. Kagan applied for a training period in the department of Pediatric Nephrology at University Hospital in Leuven, Belgium. Elena Levtchenko, MD, who is actively involved in cystinosis research and cystinosis patients' care both in Belgium and in The Netherlands, warmly accepted the candidacy of Dr. Kagan.

Mikhail Kagan will visit Europe in January 2011. During this period he will attend

[See KAGAN page 4]

Tsygin and Kagan Receive 2010 W.L. and Sophia Hobbs Humanitarian Award

By Valerie Hotz

Prior to 1983, when parents were informed their child had cystinosis, there was nowhere to turn. No patient support group existed to serve the needs of cystinosis patients and their families. As a result of the industry and compassion of W. L. and Sophia Hobbs, their daughter Jean Hobbs-Hotz and the support of many other individuals, the Cystinosis Foundation was organized and officially registered as a 501 (C) (3) nonprofit organization with the State of California in 1983. W.L. and Sophia shared the belief that every individual has a responsibility to give back to society, each according to his or her own means, and that by working together, it is possible to help alleviate the hardships and challenges in this world. They shared agape love, a passionate committed to the well-being of others.

Alexey Tsygin, MD and Mikhail Kagan, MD receive the Hobbs Humanitarian Award.

The 2010 W.L. and Sophia Hobbs Humanitarian Award recognizes the contributions of Alexey Tsygin, MD, Chairman of Pediatric Nephrology department in Russian Academy Research Center for Children's Health in Moscow, and Mikhail Kagan, MD, pediatric nephrologist at Orenburg Regional Children's Hospital, were the first to diagnose cystinosis in Russia in 2007. At one time the medical community in Russia believed that cystinosis did not exist there. However, due to the commitment and industry of Dr. Tsygin and Dr. Kagan, the first child was officially diagnosed in 2007. In the past two and a half years, an additional seven children have been diagnosed with this rare disease. There remains the challenge of getting Cystagon, the only treatment for cystinosis, approved by Russian authorities for distribution to the patients. Dr. Tysgin and Dr. Kagan are working on this crucial detail as well.

The W.L. and Sophia Hobbs Humanitarian Award may be awarded to any member of our community, recognizing the contributions of an individual that goes above and beyond. It is with deep gratitude and admiration the 2010 W.L. and Sophia Hobbs Humanitarian Award is presented to Dr. Kagan and Dr. Tsygin.

◆ PRESIDENT from page 2

taken place that year – both happy and sad - and look to the future when celebrating New Year's Day on January 1. I hope you will pause with family and friends to celebrate both Old Year's Day as well as New Year's Day and continue to live each and every moment to the fullest. We are together on this journey and I am reminded of John Donne's reflections written in 1624:

No man is an island entire of itself; every man
is a piece of the continent, a part of the main;
if a clod be washed away by the sea, Europe is the less,
as well as if a promontory were, as well as if
a manor of thy friends or of thine own were;
any man's death diminishes me, because I am involved in Mankind;
and therefore never send to know for whom the bell tolls;
It tolls for thee.

We wish you and your loved ones a happy holiday and special moments together.

With warm regards,

Jean Hobbs-Hotz,
President and Founder
Cystinosis Foundation

◆ KAGAN from page 3

four cystinosis outpatient's clinics both for children and adults in Belgium and in The Netherlands. Additionally he will get acquainted with laboratory techniques for studying the disease mechanism of cystinosis, and in performing biochemical and genetic diagnostic tests. After returning back to Russia, Dr. Kagan will spread his experience among Russian physicians to further enhance patient care.

The Cystinosis Foundation supports this project as part of its mission to improve cystinosis care all over the worl

Annual Midwest Family Gathering

By Doretta Hoffman

Nine families attended the annual reunion in the Midwest.

The Midwest family gathering was held on Friday, July 23 through Sunday, July 25 in Story City, Iowa. Don, Doretta, Jon & Andrew Hoffmann served as the host family. We had nine families in attendance, which included eight individuals with cystinosis (children and adults). The Hoffmann's welcomed the families with a welcome package and a gift to remind everyone to please be an organ donor.

On Friday night everyone was at the Hoffmann's house for a meal of grilled hamburger and brats. There were yard games to be played and a great game of ping pong. Later in the evening we watched an enjoyable DVD that Jon Hoffmann had put together of the past 20 years of the Midwest cystinosis families and past conferences. It brought back many good memories of past gatherings and how our cystinosis children have grown up to become adults.

Shawn and his Dad, Dan Reuter.

Saturday morning a few people in the group went for a swim in the motel pool. After shopping at the local mall, the annual family and group pictures were taken in the afternoon. Then we were then off to a ride on the 1913 Herschell-Spillman Antique Carousel where Terry Tschannen and Eric Gard decided to fit right in the Norwegian heritage and purchased the Norwegian Viking hats! After the great carousel rides it was off to the bowling alley for a game of "Crazy Bowling". Before we started we displayed the traveling trophy to be awarded to the winner and to take home with them till next year. After the "Crazy Bowling" game was over Jon Hoffmann presented the award. He said that since this was "Crazy Bowling" there was a twist and the award would be presented to person with the lowest score not the highest score. The trophy was awarded to Mikaela Gard.

After the bowling we were back to the motel to welcome the Ridgeway family, who are new to our group this year. After introducing ourselves it was back to the Hoffmann's for a meal and more of the competitive ping pong and yard games. Later in the evening we had a campfire and sat around the fire and shared details about where our children are in the cystinosis journey.

Mikaela Gard was all smiles.

Front Row - Shawn Reuter, Mikaela Gard, Bill Croce; Back Row -Lincoln Ridgeway, Weston Tschannen, David Reuter, Andrew Hoffmann, Nick Wagner.

This year we had lost one of our own cystinosis adults, Keri Hohl. Keri passed away just two weeks prior to our gathering. The Hohl family was able to come to our gathering, and the Midwest families presented them with a cement memorial garden plaque in memory of Keri. We were all able to help them with the healing process in the loss of Keri. Please re-

[See FAMILIES page 13]

Eye Problems and New Treatments

Monte A. Del Monte, M.D.

Cystinosis is a rare autosomal recessive metabolic disorder characterized by cellular accumulation of cystine crystals in many organs and tissues. Ocular complications of this disorder include photophobia (light sensitivity) as well as corneal erosions and later a form of corneal degeneration called band keratopathy with visual loss; all caused by increasing cystine crystal accumulation within the cornea. Crystal accumulation is also noted in other anterior ocular tissues including the conjunctiva, iris, ciliary body, as well as important posterior ocular structures such as the retina and optic nerve. These crystals begin to accumulate in the first months of life and their presence is helpful diagnostically during evaluation of infants for poor growth, or kidney problems. Figure 1 illustrates the ground-glass hazy appearance of the cornea in a three year old child with cystinosis who already has marked light sensitivity as well as intermittent pain and foreign body sensation, and decreased corneal sensitivity.

Crystal accumulation also occurs in the optic nerve and is associated with optic nerve edema (swelling) and increased pressure within the brain known as pseudotumor cerebri. Crystal accumulation in the retina results in retinal pigmentary retinopathy which begins with patchy peripheral depigmentation but later causes changes in the central vision area (macula) which can result in visual loss later in life.

Monte A. Del Monte, MD, Skillman Professor of Pediatric Opthalmology, University of Michigan,Kellogg Eye Center, Ann Arbor, Michigan

The development of oral cysteamine therapy as well as successful kidney transplantation has significantly changed the natural history of this disease. However, with increased longevity, ocular symptoms and complications have become more clinically significant, especially in patients who do not take their recommended oral cysteamine treatment regularly and from an early age. Large clinical studies by William Gahl, MD and others at the National Institutes of Health (NIH) have clearly demonstrated improvement in posterior ocular signs and symptoms such as retinopathy in patients who fully comply with oral cysteamine therapy. But oral cysteamine does not reach the cornea well and so does not eliminate the corneal cystine crystals or the resulting severe light sensitivity and corneal irritation.

To treat the corneal problem, the NIH developed a formula for topical cysteamine eye drops that were shown to be safe and effective in several clinical trials. The NIH protocol eye drops clearly dissolve the corneal crystals in young and older patients to reduce or eliminate the photophobia, blepharospasm (frequent blinking), and eye pain caused by cystine crystal corneal irritation.

Figure 2 shows the marked corneal crystal reduction and clearing in young and older patients treated with the drop. But, unfortunately, although the NIH drug protocol works well, lifelong continued compliance is difficult requiring frequent, 10-12 x/day, dosing with drops that are unstable at room temperature, requiring them to be shipped and stored frozen. By supporting a large clinical trial in which I was involved, Sigma-Tau Pharmaceuticals attempted to develop an improved eye drop formulation that would have been more stable and easier to use. But the new formula proved to be less effective than the original NIH protocol. Fortunately, however, Sigma-Tau has made the investment to bring the original NIH drops to market in the United States and just recently obtained scientific approval from the U.S. Food and Drug Administration, now only awaiting approval of a manufacturing plant to produce the drug for distribution. We are all looking forward to this exciting development that will improve local access to this eye drop treatment for cystinosis patients.

Finally, at the recent 6th International Cystinosis Congress meeting in Lignano Sabbiadoro, Italy, we heard from many outstanding international cystinosis researchers and clinicians about the excit-

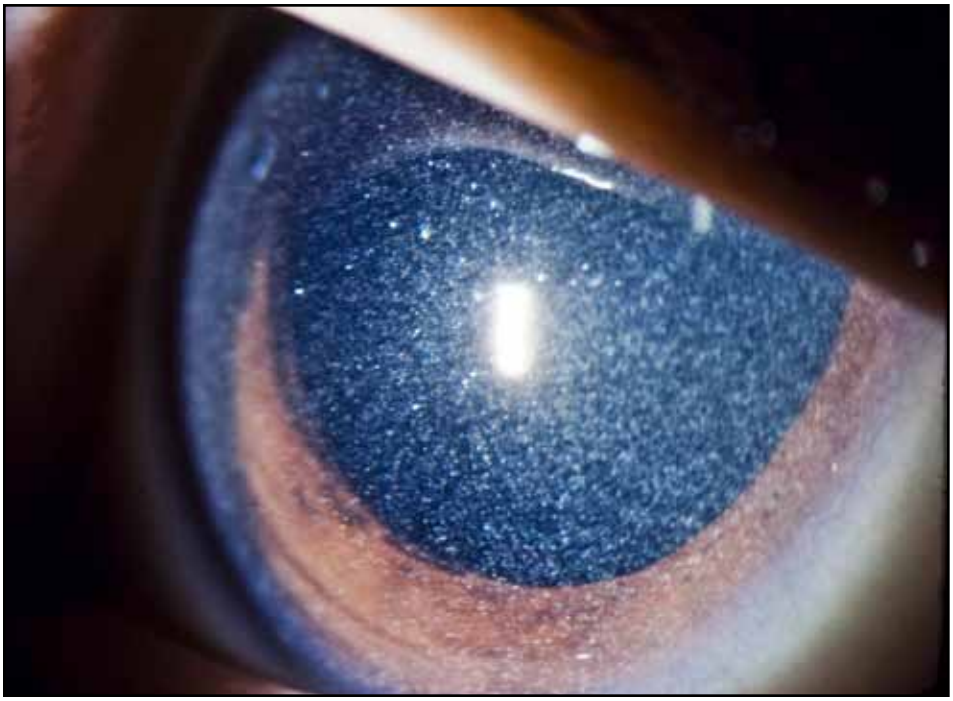


Figure 1.

[See EYES page 21]

Outstanding Long Term Patient Benefits of Steroid-Free Immunosuppression in Pediatric Renal Transplants

Li Li, MD · Minnie Sarwal, MD
Nephrology Division, Pediatric Department, Stanford University

Although corticosteroid usage has been a mainstay of transplant maintenance immunosuppression to prevent and also limit episodes of acute rejection for over fifty years, the related side effects are major causes of morbidity, including hyperlipidemia, hypertension, weight gain, diabetes mellitus, and growth retardation, especially in children. The steroid-free immunosuppression protocol developed in 1999 in pediatric kidney transplantation at Stanford University (*Sarwal M, Transplantation, 2001, 2003*) has confirmed early patient and graft benefits at one year post-transplantation in un-sensitized infants and older children. Subsequent results from a seven year long-term follow-up analysis of 129 consecutive steroid-free pediatric recipients at Stanford University confirmed the excellent benefits on low acute rejection, dramatic catch up growth and reduced cardiovascular risk profile (*Li L., Sarwal M., Am J Transplant 2008*). The first author of the latter study, Dr. Li Li, MD., MS. received a Young Investigator Award from the American Transplant Congress in 2008 for this innovative research.

"Subsequent results from a seven year long-term follow up analysis of 129 consecutive steroid-free pediatric recipients at Stanford University confirmed excellent benefits of low acute rejection, dramatic catch up growth and reduced cardiovascular risk profile."

Li L., Sarwal M., Am J Transplant 2008

Li Li, MD.

Minnie Sarwal, MD.

Efficacy and safety of Thymoglobulin induction as an alternative approach for extended Daclizumab induction

The current steroid-free protocol developed at Stanford University utilizes tacrolimus, mycophenolate mofetil (MMF) and extended daclizumab induction for 6 months for a total 10 mg/kg for steroid-free (versus 5 mg/kg for the standard 8 weeks daclizumab induction used in the steroid-based comparative group). Steroid-free recipients received no steroids except as an option for the treatment of acute rejection. All patients had protocol biopsies at 3, 6, 12, and 24 months after transplantation for monitoring graft dysfunction. The first 3 cystinosis patients who received a steroid-free transplant at Stanford have had excellent graft outcomes and significant benefits with catch up growth. For a disease that has its inherent problems with growth failure, the restoration of normal renal function and the lack of any growth suppression from chronic steroid use (the standard of care in most other transplant centers), provides a significant benefit to this patient group. In addition, the other side effects of steroids such as dyspepsia and heartburn can be synergistic with Cystagon side effects and can be avoided.

With the withdrawal of Daclizumab from the US market by Roche Inc., the steroid-free protocol has been adapted to one of rapid steroid withdrawal with a 3 days steroid exposure and Thymoglobulin induction (a drug that depletes T cells in the blood). Our recent published study (*Li L., Sarwal M., Transplantation, 2010*) has shown that Thymoglobulin appears to be a safe and effective alternative induction strategy in patients for steroid-free protocol in pediatric renal transplantation and could replace the standard of care steroid-free protocol with Daclizumab in both low risk and high risk sensitized patients.

The following data for steroid-free patients is presented from the Daclizumab steroid-free cohort, with 7 year follow-up, comparing outcomes to patients on steroid-based immunosuppression with tacrolimus and MMF and 2 months of Daclizumab induction.

Improved Graft Survival

Death-censored graft survival was significantly higher in steroid-free patients 99% than in steroid-based patients 93% ($p = 0.026$). Actual graft survival was also higher in steroid-free patient 96% versus steroid-based patients 91%.

Less Acute Rejection Episode

The overall incidence of clinical acute rejection was significantly lower in steroid-free patients, 11% compared with steroid-based patients 24% ($p=0.03$). There was no difference in the mean time to first acute rejection between two arms.

Dramatic catch-up growth rate

Catch-up linear growth, represented by Z-scores (standard deviation scores calculated from CDC growth chart), was significantly improved in the youngest recipients (<=6 years old of age) in steroid-free arm versus steroid-based arm follow-up ($p=0.01$). Surprisingly, after 4 years post-transplantation, the youngest steroid-free children showed catch-up growth rates, greater than the growth velocity in age and gender matched normal controls (based on CDC growth chart). Height benefits

[See RENAL page 19]

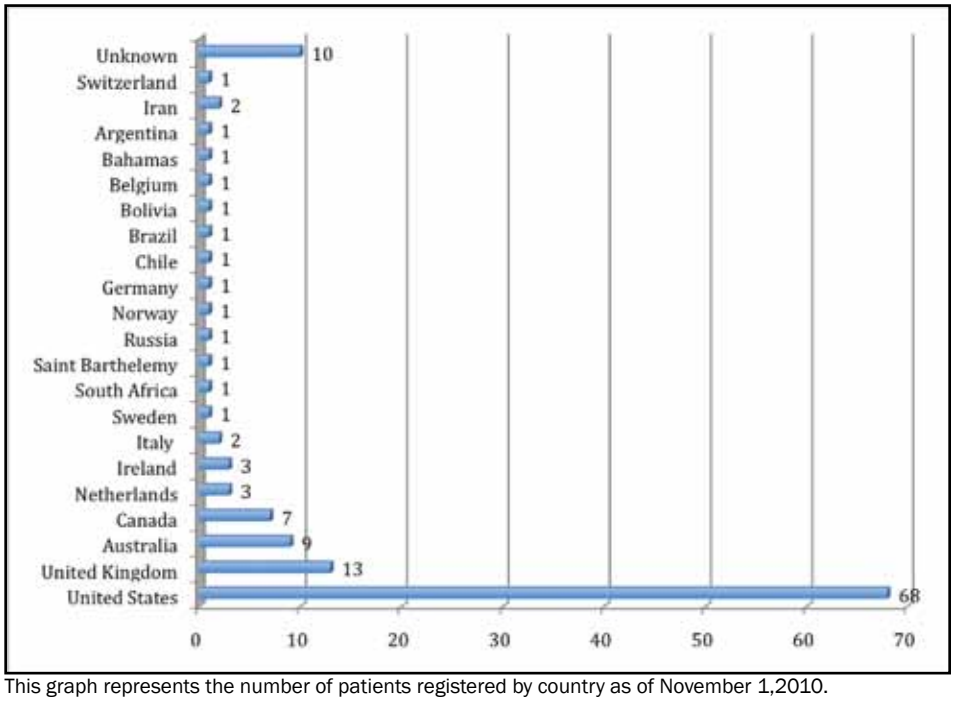
Registrations Grow to Over130 in Cure Cystinosis International Registry

By Valerie Hotz

The Cure Cystinosis International Registry (CCIR) was launched on August 25, 2010 and to date over 130 individuals with cystinosis have registered. The CCIR is the only registry created specifically for our entire global cystinosis community. It was created by medical and scientific cystinosis experts with leadership and suggestions from many international cystinosis organizations and family foundations based in the United States. The CCIR is the only secure cystinosis registry with a professional curator, ensuring complete patient privacy.

"We are concerned about some complications some adult patients are experiencing and believe registering with CCIR will help inform research scientists more fully in this regard. The Cystinosis Foundation is a committed partner with the Cystinosis Research Foundation and encourages every cystinosis patient to register with CCIR today by visiting the web site at www.cystinosisregistry.org. Please spread the word to others about this important resource and encourage them to register," says Jean Hobbs-Hotz, President of the Cystinosis Foundation.

The leader in launching CCIR, Nancy Stack, trustee of the Cystinosis Research Foundation, encourages parents to register their child and every adult patient to register as soon as possible. "The registry questionnaire is comprehensive, covering areas from diagnosis, medications, and transplants to quality-of-life questions. Once the questionnaire is completed, registrants will be able to compare their responses to others in the cystinosis community, providing a glimpse of how they fit into the larger community," says Stack. Once an account is created by an individual, he or she must complete their registration within 60 days, otherwise the account lapses. This feature has been instituted in order to encourage individuals to complete their registration in a timely manner.



This graph represents the number of patients registered by country as of November 1,2010. Registrations increased to 145 by November 15.

Countries with a dark green shade have the highest number of registrants, while those with the lighter shade of green have 1 registrant as of November 2010.

In order to accelerate the research process, participation from every person with cystinosis is crucial. The data will help scientists better understand this extremely rare disease, of which it is estimated approximately 2,000 people in the world are coping with today. "Participation in the registry will help speed up the recruitment process for clinical trials and help facilitate and expedite clinical trials. It is our hope the registry will lead to advances in improved treatments," explains Stack. The site also provides information about clinical trials. It features an "Ask an Expert" section where patients may ask personal questions that are answered promptly by a medical professional knowledgeable about cystinosis, with confidence that their privacy will be maintained.

To date, 145 patients living in 20 different countries have registered with CCIR. In addition, more than 15 researchers

[See REGISTRATION page 16]

Faces in the Crowd…

Christian Sproedt, Kirsty Biss, Freek Wonnink, Serena Scott, Annie Kwakkel, Trudy Glass and Mack Maxwell.

Frederik Pedersen, Adam Barnard and Kirsty Biss.

Back row: Ria Kwakkel, Trudy Glass, Evert Kwakkel and Erik Vickner; Sitting: Zach Ritchie.

Standing: Don Hammond, Valerie Hotz; Sitting: Dan and Holly Reuter.

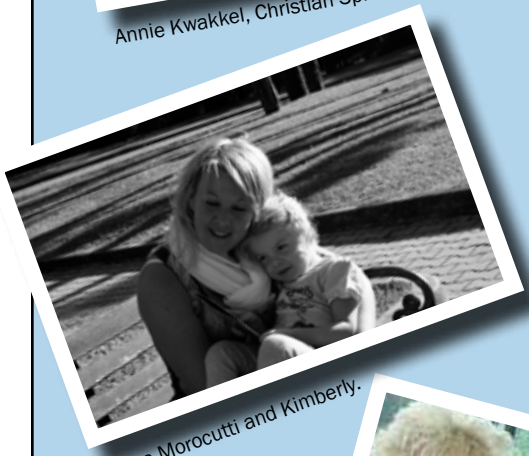
Marjolein Bos, Karen and Frank Ritchie, Erik Vickner and Zack Ritchie.

Foreground: Jan Tromp and Jan Lootens, Freek Wonnink and Katrine Boullart

Annie Kwakkel, Christian Sproedt and Joriin Visser.

Mark Tromp gives his father Jan a hug.

Jonathan Terry and share a laugh.



Laura Morocutti and Kimberly.

Tiziana Cacciatore and her daughter Alice Carmen Aronico.

Tracy Bu and Lisa Watkins.

Tom Goulsbra, Mack Maxwell and Kirsty Biss.

Ruby Watkins and Serena Scott.

Christian Sproedt, Melissa Scott, Daniel Kelly and Maria Pekli.

NOTHING IS TOO SMALL TO KNOW, AND NOTHING TOO BIG TO ATTEMPT.

Ph.D. student Lisa Frost, Professor Donald Cairns and Professor Roz Anderson, of Sunderland University.

Assoc. Professor Stephanie Cherqui, Ph.D., Scripps Research Institute.

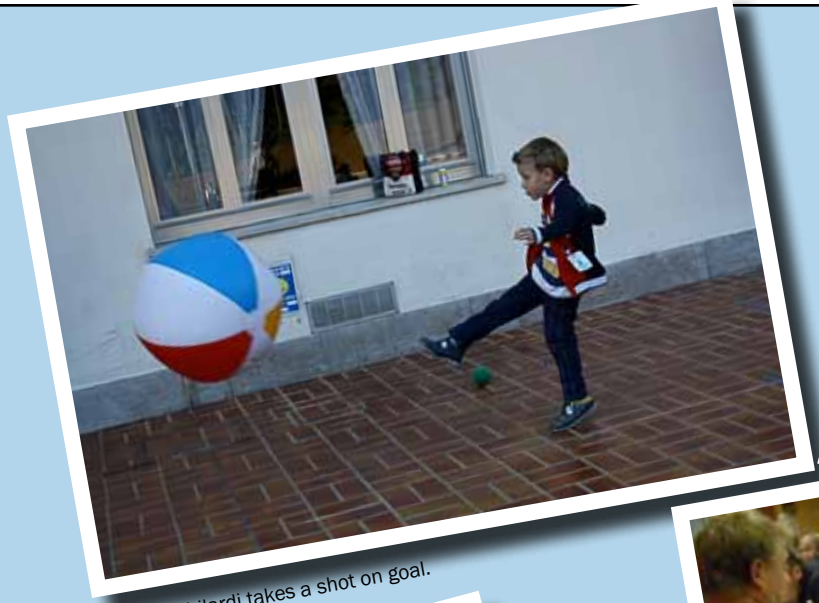
Doris Trauner, MD, Marlies Ostermann, MD and William van't Hoff, MD discuss neurological and psychological issues.

Patrick Niaudet, MD, Necker Hospital and Alexey Tsygin, MD, Russian Academy Research Center.

Professor Corinne Antignac, MD, Ph.D., Necker Hospital, Director of Inserm.

Ranjan Dohil, MD, U.C. San Diego and Rady Children's Hospital.

Elaine Schneider, Jerry Schneider, MD, surrounded by well wishers on his retirement.



Manuel Ghilardi takes a shot on goal.

Manuel Ghilardi and Ruben Mannara focus on the task at hand.

Daniel Kelly and Maria Pekli.

Francesco Manfredi and Paula Lupu have fun blowing bubbles.

Francesco and his Dad, Ivano Manfredi.

Maurizia Siliberto and her daughter Silvana Piccoli.

The Brothers Goulsbra, Josh, Tom and Adam.

Samuele Cherubini sneaks a peek.

Royalty Visits Red Cross Hospital

By Gail Daniels

Who says that you can't get 'Royal Treatment' in hospital? This was the case on Friday 18 June, 2010, when Prince William and Prince Harry visited the Red Cross War Memorial Children's Hospital in Cape Town, South Africa. This is the one and only children's hospital dedicated exclusively for children in all of southern Africa.

The main reason for their visit to the Red Cross Hospital was to spend time with patients and their families or caregivers in the Specialist Medical Ward E2 and to announce the Public Launch of the Children's Hospital Trust's fundraising campaign to develop a Surgical Skills Training Centre at the Red Cross War Memorial Children's Hospital.

Ward E2 is a renal, neurological, gastroenterology and liver facility. This is also where eight of the sixteen known cases of cystinosis in South Africa are treated.

Priscilla Windvoel is a 9 year-old girl who was diagnosed with cystinosis at the age of two years. Her mother, Carol, noticed that she had difficulty walking and took her to Red Cross Hospital. After a series of tests she was diagnosed with cystinosis. For little Priscilla this was indeed a magical moment as she looked at Prince Harry with broad smiles. She was given a photo of herself with Prince Harry and she treasures her precious photograph of the Prince.

Kamvalethu Jafta, a 3 year-old with cystinosis, was also visited by Prince Harry. Kamvalethu and his mom, Tembi, come from the Eastern Cape and have made the nine to twelve hours journey to Red Cross Hospital every 3 months since Kamvalethu was diagnosed at the age of one. Kamvalethu's hospitalizations are very challenging for Tembi, because she has to stay there with him, away from family and friends and often it is as long as three months at a time. She says that it is worth taking the trip because she knows that he receives good treatment.

The cystinosis community, especially in the Cape, is very grateful for the excellent care they receive from the team of nephrologists at Red Cross War Memorial Children's Hospital and to add 'Royal Treatment" to that is just fine. The South Africa Cystinosis Support Group is working to connect cystinosis families to one another and helping to remove the feelings of isolation that patients with a chronic, rare disease often experience.

HRH Prince Harry visits with 3 year-old Kamvalethu Jafta and his mother, Tembi.

HRH Prince Harry and HRH Prince William visit with children at Red Cross Hospital, Cape Town S.A. Prince Harry laughs with Priscilla Windoel.

◆ FAMILIES from page 5

member to keep the Hohl family in your prayers in the days ahead.

The weekend was wrapped up with breakfast and frozen fruit cups and saying our fond farewells till we meet again. Robert and Sylvia Douglas family will be our host family for the 2011 gathering.

Metabolic Nephropathic Cystinosis

By Molly Kidd

Let me start with a story, my brother Adam was born on June 16th 1985. He was a perfectly healthy baby boy weighing five pounds ten ounces. He ate so much and so quickly that his nickname was porker boy (or P.B. for short). We just couldn't shovel in the food fast enough.

By the summer of 1986 this all began to change. When Adam turned 14 months old my mother started to notice that something was just not right with him. His never ending appetite diminished and in its place came an unquenchable thirst. The sun bothered his eyes, he was constantly urinating, and he threw up quite often.

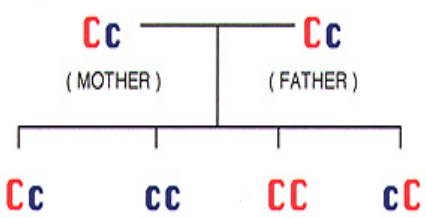
Over the next few months many trips were made to the pediatrician's office. Each time they would look Adam over and send them home with the same diagnosis, "Nothing's wrong," and "Everyone grows differently, relax". Well, "Relax" was just what my mom did not do. My brother had stopped walking and he lost 4 pounds between 1 year old and 16 months. He was no longer thriving as a small child should.

Late one December night my mom was changing Adams diapers In front of the TV when she happened to look up and see a commercial about starving children in Africa. She suddenly noticed that Adams appearance was quite similar to the starving children on the television. He had a little Buddha belly, skinny legs that showed all the glands and his chest was so thin you could count the ribs and see his little heart beat. My mother now knew that Adam was suffering from malnutrition.

The next day Adam and my mother once again went to the doctor where she demanded a test for diabetes. The test came back negative but it did show something was wrong. Now the question was "What was it"? Adam's tests showed signs of a metabolic abnormality. That was when my mom found a doctor in Oakland, California who helped diagnosis Adam. My parents were happy to have a diagnosis but what exactly was cystinosis? My family began a journey learning about, learning to cope with, and learning to live with a disease called cystinosis.

Cystinosis is a rare genetic inherited disease. The full name is actually metabolic nephrogenic cystinosis. How does a child inherit cystinosis? All people have 46 chromosomes, two sets of 23, in their cells. A chromosome can be seen as a string of beads, each bead representing a gene. Each gene controls a different function in the body. Everyone has several abnormal genes which can cause a variety of diseases. But if the other gene on the chromosome is normal, the defective gene is called recessive. The person with the recessive gene paired with a normal gene is called a carrier.

How then can a recessive gene cause a disease? The child must inherit the recessive gene from each parent to have the disease. The chances are one in four that the child of two people who are carriers will be entirely normal (have two normal genes); two in four that he will be a carrier (have one normal and one abnormal gene),



and one in four that he will have the disease (have two abnormal recessive genes).

C = normal gene / non-cystinosis gene
c = recessive gene /cystinosis gene

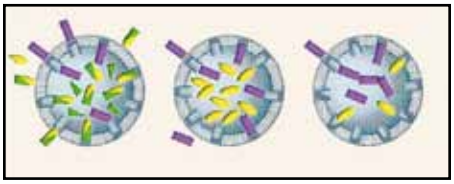
Cc = carrier / heterozygote / no cystinosis
CC = non carrier / no cystinosis
cc = cystinosis

If two people who are carriers for the cystinosis gene have a child there is a :

- 1 in 4 chance the child will have cystinosis (**cc**)
- 2 in 4 chance the child will not have cystinosis, but will be a carrier (**Cc**)
- 1 in 4 chance the child will not have cystinosis or be a carrier (**CC**)

Now that I have explained how a child gets cystinosis, I will tell you what cystinosis is. It is believed about 2000 people in the world have this disorder. An orphan disease is any disease that affects less than 200,000 people.

Cystinosis is caused when a substance called cystine builds up in the body tissue. Cystine is an amino acid, one of the building blocks for proteins. It is a necessary substance in our bodies, and it is in all the food we eat. A person with cystinosis has an excess amount of cystine. In a person



with cystinosis the cystine enters the cells but cannot get out. Cystine collects in all the body tissues but some organs are more affected by it than others.

Diagram 1 shows a healthy cell where the cystine enters the cell and then releases what it does not need.

Diagram 2 shows the cell of an individual with cystinosis. The cystine enters the cell and it is trapped within the cell and cannot escape.

Diagram 3 shows the cell of a cystinosis patient who is taking the only treatment for cystinosis. This treatment is Cystagon. The Cystagon binds to the cystine and allows it to be released from the cell.

The kidney is one of the organs that seem to be damaged the most. The kidney has two main functions, to filter the waste in our body and to reabsorb the water and nutrients our body needs to maintain a chemical balance. In a normal kidney waste products are filtered from the blood. Then water plus salt, potassium, bicarbonate, phosphorous, sugar and some protein, along with the waste products pass into the kidney tubules. The tubules then reabsorb most of the water and the salt, potassium, bicarbonate, phosphorus, sugar and protein, and return them to the blood. The waste products and a small amount of the filtered water pass down through the tubules and into the bladder as urine.

Early in the disease the tubules lose their ability to reabsorb the water and other substances. It is because water is being lost in the urine instead of being reabsorbed that children with cystinosis have to drink and urinate so often. The amount of sugar and protein that is lost in the urine is actually

[See METABOLIC page 15]

◆ METABOLIC from page 14

very small, and no replacements are needed. However, the water, salt, potassium, bicarbonate and phosphorus have to be replaced as these are very important.

The second job of the kidney, filtering waste from the blood is also damaged. This is a gradual process. The damaged filter allows waste products to accumulate in the blood and remain there. When this happens, the cystinosis patient has kidney failure. The treatment for kidney failure is dialysis and/or kidney transplant.

Another organ that is found to be damaged by the cystine crystals is the eye. Cystine can actually be seen in the eye using a slit lamp. This causes the children to feel like there is sand in their eyes. These children are very sensitive to light and will wear dark glasses and a hat. They now have eye drops for these children that must be used every hour during the day.

There are three simple ways of diagnosing cystinosis. The first way would be to take the measurement of leukocyte cystine content. Another way would be to examine the amount of crystals in the eyes by using a slit lamp. And the third way would be to take a biopsy of the kidney.

The treatment for cystinosis is a complicated process. The first treatment is to correct with medicine the abnormalities in sodium, potassium, bicarbonate, calcium, and phosphorus that may be present when the child is first diagnosed. This can take a while to correct, because it is hard to find the right dosage for each medication. The children need regular blood tests to check all of the levels needed to balance their blood chemistries so their electrolytes are as normal as possible.

Of all the medications, Cystagon, potassium and bicarbonate are probably the most important to take regularly and on time. Cystagon must be taken every six hours, around the clock. Not taking the medicines may be very dangerous. Calcium, phosphorus and Vitamin D need to be balanced in the bones as well as in the blood. If these are not balanced, it can lead to a disease called rickets. Rickets is very rare for people in the United States. Rickets is caused by lack of nutrients in the bones. We mainly see rickets in children that are malnourished and starving. The medicines not balanced can also lead to tetany. A child with tetany can develop cramping in the muscles of the legs, hands or feet. This can become very severe and lead to convulsions.

Some of the medications required for cystinosis patients are:

Polycitra: Acts like as a base to neutralize his acid body solution.

Calcium & Vitamin D: Aid in bone strength and teeth development.

Nuetra-Phos: Keeps an adequate amount of phosphorous in the body.

Thyroid: A medication that takes the place of the inactive thyroid gland. Without this hormone, the child would cease to grow.

Cystagon®: Aids in the breakdown of cystine in the cells. Without this, none of the other nutrients would be absorbed.

Water is one of the main concerns in a child with cystinosis. As the kidneys can't reabsorb water after it is filtered from the blood, the children have to urinate and drink all the time. They need to have liquids available all the time, including throughout the night. A normal kidney keeps a constant level of water in the body by regulating the amount of urine produced. In a normal body the more you drink the more you urinate. If a person drinks more, then they urinate more. If we drink less the kidney acts like a valve and makes less urine keeping the amount of water in the body constant. In cystinosis, the kidney is like a tank without a valve. If a cystinosis patient drinks less, the amount of urine remains high while the amount of water in the body falls, causing dehydration. Dehydration can be a serious problem in cystinosis.

Cystinotic children will find a way to drink whether it is drinking out of toilet bowls, using a dog dish to get water, or even licking the dew from the windows. One of the problems with drinking this much water is that the child will drink to the point of vomiting and then drink some more. Adam drank 17 liters of water per day and urinated most of it out. Consequently, he didn't eat enough to gain weight. At 5 years of age Adam still weighted only twenty-four pounds. Adam and many children with cystinosis have a feeding tube (gastrostomy) put in. This tube is put in the stomach and it opens on the outside of their body. At night this allows the children to get nutrition while they are sleeping. This tube also allows them to get medications when the child is ill.

Another trait of these children is their unusual dietary habits. These children don't seem to have a liking for sweets. My brother's idea of a great meal was a tomato, a pickle, a big blob of ketchup, and some mustard. Most of the children like spicy food: Mexican, Chinese, and Italian. The reason why most cystinotic children

[See METABOLIC 2 page 16]

Author Molly Kidd is 12 years-old and learning how to captain her ship.

Life With Cystinosis

By Adam Kidd

My name is Adam Kidd, I am almost ten years old and I live in Santa Rosa, California. I have a very rare disease called Metabolic Nephropathic Cystinosis. This is a very rare disease with only two hundred children in the United States who are affected. I have been living with cystinosis since I was eighteen months old. I have a couple of friends with the disease that live in the United States. Lyndsie lives in Kansas and Kevin lives in Hawaii. Cystinosis is a very hard disease to understand and even the doctors have a hard time understanding it. I'm going to tell you about some of the good and bad things of living with cystinosis.

One of the good things is I am able to meet really nice people. I have a really great doctor who I really love. Because I am kind of special I get more attention. Since I have cystinosis my mom can't work anymore, so she is home with us every day of our lives. I do home school and I get to do archery and ice-skating for P.E. I get out of school very early sometimes and get to play games to learn.

The late Adam Kidd.

Some of the bad things about my disease are whenever I get sick I have to go to the hospital, usually for a long time. They always take a lot of blood tests and put in a lot of I.V.'s. I always have to drink really nasty medicines in four cups every day and take a ton of pills. I am really short because my body doesn't absorb the food I eat. I drink almost seventeen liters of water a day. At night, I am hooked up to a feeding tube to give me more calories and medicine. Sometimes, I have a hard time on a hot sunny day because the sun hurts my eyes and I do not sweat.

It is never any fun to have a disease, but I have a good life. I have a Mom, Dad and Grammie who love me. A sister and a brother I love. I also have three dogs, a cat, and a bird.

I am a very happy boy.

Editor's Note: Adam wrote this essay in 1995, when he was 10 years old. He passed away at the age of 12.

◆ METABOLIC 2 from page 15

have a craving for this kind of food is because they are replacing the nutrients their bodies are lacking such as, potassium, salt, and protein.

At this time cystinosis is not a curable disease.

Cystinosis Research Foundation's primary focus is to find a cure for cystinosis. In April 2009, we met with a team of CRF-funded scientists from Scripps Research Institute in La Jolla, California. They presented us with a plan that will lead to clinical trials in children with cystinosis within the next five years. The clinical trials will test new gene therapies that researchers believe could lead to a cure for cystinosis. This plan holds the first real promise that a cure for cystinosis is a possibility.

My brother Adam Kevin Kidd lost his battle with cystinosis when he was 12 years old.

The doctors believe that the cystine crystals blocked the electrical current in Adam's heart. I was born 5 months after my brother died. From the stories and pictures, I feel as if I knew him. Everyone has shared stories of Adam with me and they all say he lived life to the fullest.

A Note from the Author

My name is Molly Amanda Kidd. I am twelve years old and currently in 7th grade attending Washington Virtual Academy in Santa Rosa, California. Last year as my end of the year writing assignment I needed to write a research paper. I chose to write my report on cystinosis and read many articles by authorities including Dr. William A. Gahl, Dr. Jerry A. Schneider, Dr. Patrick Niaudet, Dr. Jess Thoene, as well as the Cystinosis Foundation's web site and publication, "Cystinosis", the Cystinosis Research Foundation web site and personal interviews with my mother, Susan Kidd.

The inspiration for this paper was my brother Adam Kevin Kidd. My brother was diagnosed with cystinosis at the age of two and he lost his battle with this disease when he was twelve. I was born 5 months after my brother earned his wings in June of 1998. My middle name Amanda was chosen because the letters of my brother's name are included in Amanda. Adam was a very outgoing, high spirited, fun loving boy whom everyone loved and cherished. Even though he left too soon I know that he is in heaven partying with the angels, and doing some of his favorite things. I know he is doing what he does best, being himself.

I work really hard in school so that I can be a Family Nurse Practitioner one day. If I am really lucky I would like to travel around the world with my best friend Eileen. I live each day trying to be the best I can be, wanting my brother to know that I am following in his footsteps living life to the fullest.

◆ REGISTRATION from page 7

have queried the registry, seeking information about what it is like to have cystinosis. The registry provides patients with an opportunity to be heard by answering these very disease specific questions. The CCIR is already making an impact, as it is currently being accessed by 11 leading researchers who specialize in the field of cystinosis. The CCIR web site is located at www.cystinosisregistry.org.

In Loving Memory of Keri Lee Hohl 1986-2010

By Joan Hohl

Keri was born at 2 a.m. on November 17, 1986, to Lonnie and Joan Hohl of Sioux City, Iowa, weighing in at 7 lb. 6 oz. She had big blue eyes, very fine blond hair, and a temper when she did not get fed quick enough! Keri seemed like a typical infant, except for her ability to literally 'guzzle' down her formula in a short time. Her mother, Joan, was (and continues to be) employed as an Early Childhood school social worker - so was always checking Keri's development against the expectations. All was going well until about age 15 months. At this time Keri got chicken pox followed by strep throat - and from that point on seemed 'different.' She was often fussy, did not want to eat, and drank large amounts of water. Occasionally she would vomit a bucket of liquids. It soon became apparent that Keri was not growing - and in fact was losing weight. Something was definitely very wrong with Keri. The family physician knew he needed to refer Keri to some type of specialist in Omaha, Nebraska, which was 100 miles away.

After a misdiagnosis and several weeks of a constantly fussy toddler - Keri was diagnosed with cystinosis through a biopsy done during exploratory stomach surgery on her 2nd birthday. We were told that Keri had significant kidney damage, and that there was no 'real treatment' available for cystinosis. The most that could be done would be to replace the minerals and salts which were causing Keri's electrolytes to be out of balance (which in turn caused the vomiting). There was also mention of a research project to treat cystinosis, going on at the National Institutes of Health (NIH) in Bethesda, Maryland. We were given the option of checking into the project if we wished. It was suggested that 'we need not rush out to NIH'. Needless to say, by January 1989, Keri, Mom, and Dad were on a plane heading to the NIH. It was there we met Dr. Bill Gahl. He became our ultimate guide for living with and treating cystinosis. Keri was at that time signed up for 'an experimental treatment' for cystinosis, with a drug called phosphocysteamine.

This not only began the treatment for cystinosis, but also the desire to create a network of Mid-West families. Another family and ours worked to create an annual 'Mid-West Family Gathering' which we continue to hold each summer. It is sort of like a 'family reunion.' Keri always looked forward to our gatherings with our 'cystinosis family'; and her younger sister, Kasey, would ask: "When are we going to see our cystinosis cousins?" We never knew how much this network would mean to us - until this difficult time of Keri's passing.

The late Keri Hohl.

Time passed quickly with all the trips to the local hospital for blood work, to the University of Nebraska Medical Center to see her pediatric nephrologists, and to the NIH for experimental treatment. There were many ups and downs of cystinosis. Keri was of course 'short for her age' and always thought to be younger than her actual age. But that did not stop her from loving the game of basketball! Her dad and - whoever else could be drafted - often shot hoops with Keri on our driveway. Michael Jordan was Keri's basketball hero - and as an elementary student, we signed her up to play basketball on a team through the local Salvation Army. When she was in middle school and high school, Keri played basketball through a program called 'Upward Basketball'.

It was also in elementary school that Keri began to take a rather serious interest in learning to cook. It was not long before she would be volunteering to cook the evening meal at our house - which was great by me! Keri began experimenting with a variety of spices and herbs and particularly ground beef. She became an avid fan of Rachel Ray. As she began planning for post-high school training and employment, the culinary arts rose to the top of the list.

Keri graduated from high school in 2006. In spite of cystinosis, learning disabilities, and numerous medical appointments, Keri was finally graduating and we celebrated. With some guidance from her teachers and her parents, Keri was going to attend Iowa Western Community College (IWCC) in Council Bluffs, Iowa and study Culinary Arts. Unfortunately, as the summer progressed, so did her decline in kidney function. By July, 2006 we realized it would not be long before Keri needed either a kidney transplant or dialysis. Attending IWCC to study Culinary Arts had to be postponed. Keri went on dialysis in September, 2006. Unfortunately, neither of her parents passed through the rigorous testing to be the kidney donor for Keri. So with all the recipient testing done, Keri was placed on the 'kidney transplant list' via the University of Nebraska Medical Center/Lied Transplant Center.

On September 26, 2008, after 2 phone calls within 24 hours, Keri was finally on her way to Omaha to receive a kidney transplant. The surgery took all of 1 ½ hours and the match was made in heaven. In just 3 days Keri had normal kidney function for the first time in her life!! So with her newfound health, Keri was re-registered and attending IWCC and the Culinary Arts Program in Council Bluffs, Iowa in the fall of 2009. We were very hopeful for Keri's success, but knew there were more challenges. Between Keri's health issues and the lack of educational accommodations on the part of IWCC—she returned home in Dec. 2009. She soon settled back into being home, with the hope of trying the Culinary Arts Program at Iowa Western Community College again in the fall of 2010.

It was the summer of 2010, and keeping up regular blood work at the local hospital to continually check anti-rejection levels was still part of Keri's life, a part she was

[See MEMORY page 18]

◆ REPORT from page 1

and conference costs. One of the aims of this conference was to draw attention to the young adults with cystinosis, and explore not only the problems they face in daily life but also to share their personal experiences with each other. Special in this field, this conference is truly unique and has been a huge success. It doesn't make a great difference where you live in the world: Germany, France, United Kingdom, The Netherlands , Denmark , South Africa, Italy, Ireland, Australia, or…. a lot of the experiences are the same.

Next to all the general presentations with information about cystinosis and the treatment, a lot of very promising and hopeful presentations were delivered by researchers about future treatments. I really want to mention the time-release cysteamine and the trials with this medicine which has started in the USA and in Europe (one clinical trial site is located in The Netherlands!). If this treatment is successful, all the families dealing with cystinosis will have back their night's rest!!

The bone marrow transplantation in combination with gene therapy reported by Stephanie Cherqui, Ph.D., Associate Professor at Scripps Research Institute, is important for the distant future and many share the hope to finally find a cure. The research to decrease corticosteroids, or entirely avoid their use in therapy after transplantation is encouraging. The launch of the Cure Cystinosis International Registry, an online patient registry dedicated solely to cystinosis patients, is a big step forward. Learning all this new and important information filled our hearts with hope.

In this article I really want to add one of my favorite personal experiences during this conference which was meeting with Gail Daniels. Gail has founded the Cystinosis Foundation South Africa and her stories about the children with cystinosis in her country are so impressive. They often must deal with extreme poverty, in addition to the lack of information. As many as eleven different languages are spoken in the region, adding additional challenges to Gail's work as the president of the support group. To talk with Gail made me silent.

During this conference we have said farewell to Dr. Jerry Schneider on the occasion of his official retirement. His work and research will always be the basis of what we know about cystinosis. He is a dedicated and compassionate doctor who is able to understand the families in their struggle in daily life. But it is heartwarming to see the next generation of doctors and researchers working hard to fully understand cystinosis and find the best treatment for our loved ones.

You will understand that we from The Netherlands look forward to 2012 to attend the 7th International Cystinosis Conference.

◆ FIRST from page 1

pictures in the newsletters.

The sessions were hectic, so much information that my head was full, but I kept on listening, trying to soak up as much as I could. After years and years of reading and gathering information on the website, newsletters and publications on my own, now for the first time, I was hearing it from the professionals. Now it was validated and somehow it has all become so real, so emphatic.

It is difficult to describe in words the emotions as we observed each professional getting onto the platform, sharing the effects of cystinosis in his or her area of specialty with such dedication, passion and gentleness. I appreciated the effort they extended sharing the information in a clear and direct manner; offering guidelines and tips as well as hope that comes with compliance. I definitely had mixed feelings and a sense of being overwhelmed, yet much encouraged as I sat listening, surrounded by a community of cystinosis families from across the world, all ready to support one another.

I was so impressed by these doctors. Their perseverance and diligence as they continue to research and find better treatments to improve the quality of life of their patients is remarkable.

For our daughter, Danielle, age 17, with cystinosis, this was another experience altogether. We had our own fears of her fitting in and making friends as she is a very shy person. Instead, we were surprised as to how quickly and easily she made friends. She had so much fun and enjoyed the time with Uncle Don and the group. She has learnt so much but most importantly, somehow, she has gained confidence in knowing that she is not alone and was very encouraged by the young adults and their stories.

We consider ourselves very blessed to have been able to attend this conference and also to represent not just South Africa but the continent of Africa. We are encouraged but also challenged to now do our part, which includes diligently administering the medications and caring for our children, making sure that we are complying with what the professionals have described as the best possible treatment to ensure a good quality of life.

◆ MEMORY from page 16

not fond of. On July, 6th I offered to drive her to the hospital for her blood work, and then we would go out for breakfast. My proposition worked, as we completed the blood work and then headed to Perkins. Shortly after returning home, Keri began complaining of a very sharp chest pain that would not quit. Her dad happened to come home and he took her blood pressure and pulse. They were high and racing.

After calling Keri's nurse coordinator at the transplant center, he took her up to the local emergency room. IV's were started, additional tests ordered, and Keri soon became an inpatient. Communication between the local hospital and Keri's nephrologist in Omaha over the next 24 hours determined that she should be transferred to University of Nebraska Medical Center. She was moved there by ambulance and was admitted on July 7th. The rest of Keri's stay in Omaha was an unreal blur of doctors, nurses, procedures, machines, and the unknown. By the early morning of July 8th Keri had been moved from the Cardiac Unit to the Intensive Care Unit - and then on into the Hands of God. Keri went home to a world of no pain and no more pills. She is free at last!

To My Parents, Upon My Graduation from Providence Baptist College

By Rebekah S. Palmer

Starting Bible College is a right step for a Christian high school student to make.

Finishing Bible College is a determined step towards beginning to make a difference for the cause of Christ and His future followers.

After 4 ½ years of Bible college, I look up and ask God: "Why me?" I have asked my Lord this question many times before. When I was twelve years of age, my mother donated her kidney to me so I could have a second chance at life. Six months after the transplant, I battled B-Cell lymphoma cancer with a 3 month to 3 year sentence in medical science against me. I can tell you in those times and the years before and after, my attitude of "Why me?" was not a respectful question to God Almighty.

Now, as I stand in college graduation robes, I assure you that I ask "Why me?" with the utmost awe and respect to my Maker for I know people who do not finish Bible college because of various reasons, their health included.

What explains me is not great personal character, for that needed building upon (thank you Pastor Gomez and Bro and Mrs. Hall), but what explains me is Christ and the great preparers God placed in my life who laid a strong foundation: my parents.

Rebekah S. Palmer.

I have never parented a chronically sick child; however, I would like to attempt to put you in the mindset of the mother and father who are responsible for my life.

Fanconi Syndrome (Cystinosis)

Prognosis: progressive damage to organs ending in death. Kidney transplant should be done by 9 years. Cystinosis will not affect the new kidney.

Put yourself in the mind of the young mother or in the mind of the young father. You are both 28 years old with a 5 year old girl and a 4 year old boy. You read this definition of the disease the doctors just told you that your daughter is afflicted with due to the results of a kidney biopsy and confirmed by an eye examination. What would you do for the little girl you dreamed of, desired and prayed for?

My mom decided to go all-natural. My dad decided to go right to the doctors who studied the genes and chromosomes of this disease and could pin-point from appointment number one what we needed to do for a healthy life.

We went to a naturalist doctor for a bit until my mom decided to stop running to make her daughter better and let God make her daughter better by following my dad's decision. She began listening to David Ring on Christian Radio. His story was one of seemingly terrible health circumstances yet he became a preacher of the Gospel despite his cerebral palsy.

During an especially rough day of vomiting and nausea for me, I remember my mom saying these words, "We all have a cross to carry in life. Your cross is called cystinosis. You will carry it your whole life and your choice is whether to carry it happily or grudgingly."

When mom would get frustrated from washing bed clothes every day from my mess, tired from making doctor appointments, and cranky from spending all day compiling medicines and taking care of sickness, she would look at the other kids in the hospital and praise God that I could walk, learn, and travel places. It was while looking at other children with medical illnesses that she realized God is real. God is good because she and my father were well enough to work to pay my medical bills.

My father's concerns were about the future. He began to have a better prayer life and to understand the Bible verse that claims, "In whatsoever state I am, therewith to be content." He would be amazed that though many days I was crying, I still had joy. When he was stressed, he would think about placing me with my cystinosis in God's hands.

We read in the Bible from Mark 8:36 "For what shall it profit a man, if he shall gain the whole world, and lose his own soul?" May I say that my parents have poured their soul into my life and though they have not gained the whole world, they have certainly not lost the investment they have placed in me.

Congratulations Mom and Dad on your graduation from Providence Baptist College.

◆ RENAL from page 7

were also seen in steroid-free children 6-12 years of age (p=0.01) compared with steroid-based children. These data are extremely reassuring for patients with cystinosis as it suggests that significant benefits in catch up growth can continue many years after transplantation if there is growth potential for many years after transplantation. Thus, earlier and preemptive transplantation should be strongly considered in nephropathic cystinosis.

Benefits on graft function, cardiovascular risk factors and infections

Graft function was significantly higher in steroid-free patients than in steroid-based patients (p<0.0001) regardless of recipient age.

The incidence of post-transplant diabetes was significantly lower in steroid-free arm 0.8% versus steroid-based arm 9% (p=0.01). Steroid-free patients had significantly lower requirement for anti-hypertensive agents (p<0.0001) compared with

[See RENAL 2 page 21]

Danielle Daniels: Cape Town, South Africa

Danielle is a 17 year old girl with cystinosis. She is currently studying visual arts, with photography as her practical component. She also enjoys drawing, beading, watching movies and road trips. Her best moments are spending time with family and friends.

Pearly Beach.

Courage.

Shine.

Rorman and Reuter Receive 2010-11 Deanna Lynn Potts Scholarship

By Valerie Hotz

The Cystinosis Foundation is pleased to announce Ellen Rorman and David Reuter have each been awarded a $1,000 Potts Scholarship for the 2010-2011 academic year to help them pursue their college studies. Both students achieved high school grade point averages over 3.5 and engaged in extra-curricular activities as well.

Ellen Rorman of Eagan, Minnesota, is a June graduate of Eastview High School. She enrolled at Minnesota State University at Mankato as a freshman this fall, with her sights set on entering the nursing profession. A member of the Eastview High School Speech team for two years, Ellen balanced a very busy schedule extremely well and was also a dedicated competitor on the cheerleading squad.

David Reuter graduated from St. Clair High School, St. Clair, Minnesota this past June and is a student at Bethany Lutheran College in Mankato. In addition to having a full senior course load, David enrolled in college classes at Rasmussen College. A member of the National Honor Society, he was also active in the drama club and volunteered in his local community. David is looking forward to studying art in college.

The Cystinosis Foundation congratulates Ellen Rorman and David Reuter and wishes them all the best as they follow their dreams in college.

The Power of Your Gift

For the past 28 years the Cystinosis Foundation has been a leader in spreading knowledge and providing support to families and individuals affected by cystinosis. Your tax deductible contribution supports the Cystinosis Foundation mission to educate families and medical professionals about cystinosis, to provide emotional support for those coping with this rare disease and to encourage and support research on an international basis for better treatments and a cure.

The First International Cystinosis Congress in the East Mediterranean was hosted last year in Ankara, Turkey and we have recently concluded the 6th International Cystinosis Congress in Lignano, Italy. As we continue to broaden our reach to cystinosis patients around the world, it is clear there is more work to be done to educate the international medical community. This vital effort will enable diagnosis to take place earlier in life, so a child may receive medical treatments sooner and the improved long-term outcome that results from early diagnosis.

Thanks to the generosity of our supporters over the past 28 years, tremendous progress has been made against cystinosis. Beginning in the late 1980's the Cystinosis Foundation funded vital research in the laboratories of cystinosis experts including Dr. Jerry Schneider, Dr. Jess Thoene and in the 1990's Dr. Corinne Antignac and many others. The hosting of our unique educational family conferences has contributed vastly to strengthening and expanding the international scientific community, which has accelerated advances in cystinosis research.

There is a critical need to develop educational pamphlets, brochures and DVD's for distribution to medical schools, hospitals and clinics around the world. Your gift supports this important effort to increase knowledge and helps achieve the goal of beginning treatment at the earliest possible age for young patients. We are grateful for your past support and encourage you to make a tax deductible gift to the Cystinosis Foundation today.

◆ EYES from page 6

ing future with possible new treatments for the eye complications of cystinosis. They presented new research to develop therapies and drug delivery systems that could improve and simplify treatment of the debilitating ocular complications of cystinosis. Studies were presented for discussion on techniques to improve ocular treatment using gel delivery systems, delivery of the medication in the form of a stable "pro-drug" which can be stored easily and would require less frequent dosing, that would then be activated within the ocular tissue. Evidence was also presented on gene therapy or stem cell replacement therapies that may potentially even further revolutionize the way cystinosis eye problems are treated in the future. I am encouraged that this continued research will result in new therapies which are more stable, easier to use and more long lasting.

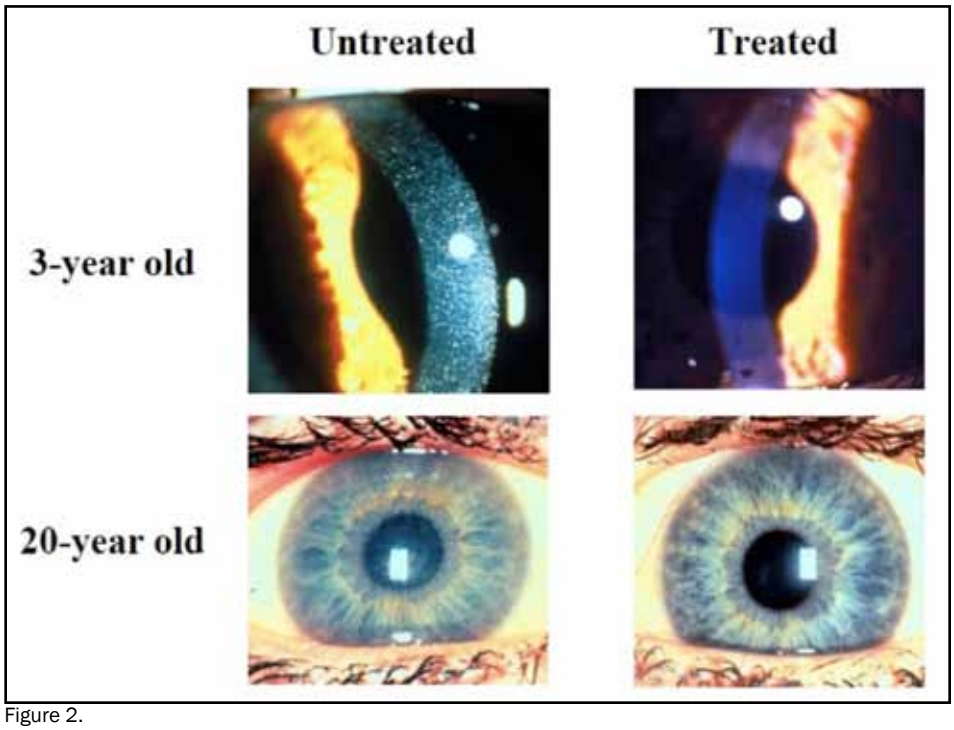


Figure 2.

◆ RENAL 2 from page 19

steroid-based patients. Serum triglyceride levels as well as serum cholesterol levels were also significantly lower in steroid-free arm compared with steroid-based arm for all time points of follow-up. In addition, steroid-free patients showed significantly smaller changes in body mass index for the first two years post-transplantation compared to steroid-based patients (p=0.02).

Subclinical EBV viremia, measured by peripheral blood quantitative PCR, was detected in 22% of steroid-free patients and 57% of steroid-based patients ($p < 0.05$). There is no difference in the incidence of subclinical CMV viremia (*Li L. Sarwal M. Pediatr Transplant 2007*).

Results Confirmed in a Prospective Multicenter Randomized Trial

A prospective controlled randomized multicenter open label trial of steroid-free immunosuppression using the Stanford protocol versus steroid-based immunosuppression has been conducted in 130 low-risk children receiving primary kidney transplants across 12 centers in the United States. Three year follow-up has been completed and analysis of the data is underway; the one year follow-up results have confirmed the benefits seen in the single center study at Stanford (*Sarwal M. Am J Transplant, Supplemental 2008*).

Deanna Lynn Potts Scholarship 2010

Application Form

(This form and the completed application may be photocopied)

Please Print Legibly or Type

FAILURE TO COMPLETE ALL RELEVANT PORTIONS OF THE APPLICATION WILL RESULT IN DISQUALIFICATION.

Name_____
Last First Middle

Permanent
Address _____

City_____ State_____ Zip code _____

Home Phone (_____)_____Email_____

Birth Date_____ Gender(___) M (___) F

Social Security No._____

Name of Current High School_____

Address _____

City_____ State_____ Zip Code_____

School Phone (_____)_____Fax(_____)_____

Principle _____

Guidance Counselor _____

Date of High School Graduation _____ Cum. GPA _____

Name of College/University/Vocational School you will attend in the fall of 2010:

Address _____

City_____ State_____ Zip Code_____

Email_____

AGREEMENTS: If I am selected as the Deanna Lynn Potts Scholarship recipient, I give permission for the Cystinosis Foundation to publicly announce my name. In doing so, I realize that I will be identified as a person with a disability. ( ) YES ( ) NO

I certify that all of the information on this application is complete and accurate to the best of my knowledge and the accompanying essay is solely my work.

(Applicant Signature) _____ Date: _____

Deanna Lynn Potts Scholarship Criteria

Deanna Lynn Potts was born with cystinosis and lived to be 27 years old. Before she died she discussed her wishes to start a scholarship fund for children with cystinosis. We know how devastating a chronic illness can have on a family emotionally, socially and financially. Children with cystinosis are living longer thanks to medical science and therefore embarking on careers. These careers require education. Education is expensive; something we do not want to deprive our children of in today's world. Due to the financially draining medical costs it might prove difficult to send a child to college. Through this fund we hope to help some of these students.

PURPOSE: To provide supplemental financial assistance to an undergraduate student diagnosed with cystinosis and enrolled in an accredited collegiate or vocational program.

SCHOLARSHIP AWARD: A $1000 Scholarship awarded annually. The award is contingent upon the winner's acceptance to an accredited college, university, or vocational program and will be payable to the educational institution to be applied to tuition, room and board.

ELIGIBILITY: Each candidate must be a current high school senior or high school graduate, who has had to postpone higher education, and has cystinosis.

APPLICATION PROCEDURE:

Applicant must submit:

Documentation/verification of cystinosis (e.g. Letter from physician.).

An official copy of high school transcript.

Two letters of recommendation from current teachers/faculty members and/or counselors regarding applicant's scholastic aptitude and personal qualifications.

An essay of 500 words. We want to know a person who played a vital role in the student's life. How? And why? The essay should be typed and double-spaced.

JUDGING CRITERIA: The essay will earn a possible 40 points and will be judged on the basis of rationale, grammar, and comprehension. Transcripts and letters of recommendation will carry a possible additional 20 points each. The Cys-

[See SCHOLARSHIP page 23]

◆ SCHOLARSHIP from page 22

tinosis Foundation Board will establish an independent judging panel to evaluate and rate applicants.

Finalists will be interviewed before selections are made.

Deadline For Application: The application and all accompanying documents must be received at the Cystinosis Foundation Office in a single, flat envelope by March 30, 2011.

Faxes or Emails Will Not Be Accepted, and applications received after this date, regardless of postmark, will be disqualified.

Preparing Application Package: Each application packet must include a completed application (original or photocopied), required documentation materials, and essays, on 8 ½" X 11" white paper. Send all materials in a single flat package. All application documents become the property of the evaluation committee.

Mail Application To:
Cystinosis Foundation Scholarship Committee
58 Miramonte Drive
Moraga, CA 94556, USA

Cystinosis Foundation Mission

The Cystinosis Foundation was established in 1983 with a mission to educate patients, families and medical professionals about cystinosis, to provide emotional support for those coping with this rare disease, and to encourage and support research for improved treatments and a cure.

Our mission is accomplished through the publication of newsletters and brochures and the hosting of unique educational family conferences that include medical professionals. In 2000, this mission was extended internationally to reach and unify cystinosis patients, wherever they live in the world.

Cystinosis Foundation Membership Application

In order for the Cystinosis Foundation to increase its resources, develop new program initiatives and continue as a strong advocate for our children and families, more members are needed. If every member of the Cystinosis Foundation recruits at least one new member, the results will speak for themselves. Membership is open to all who wish to assist the Cystinosis Foundation, a 501 (c) (3) nonprofit organization dedicated to providing education and emotional support for children and adults coping with cystinosis, as well as their families and to providing educational programs and research grants to medical professionals. Please send your donation with this completed form to: The Cystinosis Foundation, 58 Miramonte Drive, Moraga, California, 94556, U.S.A. Your gift to the Cystinosis Foundation is fully tax deductible. Our federal tax ID is 94-2927892.

(PLEASE PRINT)

Honor Circle	25,000
Patron	20,000
Lifetime	2,500
Visionary	1,000
Professional	200
Supporter	100
Family	50

Yes, I want to become a member of the Cystinosis Foundation.
Enclosed are my membership dues of $_____

No, I do not wish to become a member at this time, but I do wish to make a contribution.
Enclosed is my gift of $_____

Please accept my donation of $_____, given

In Honor of_____

In Memory of _____

____ I would like to join the cystinosis community by becoming a free member of the Cystinosis Foundation.

NAME_____

ADDRESS _____

CITY _____ STATE _____ ZIP_____

PHONE _____ E-MAIL _____

Does your employer participate in a matching gift program? Yes No

Name of employer_____

Do you belong to an organization that may be interested in a fundraising activity for the Cystinosis Foundation? Yes No
Please contact me to discuss planned giving options. Yes No

Visit our new Message Forum on our website at www.cystinosisfoundation.org

Cystinosis Foundation
58 Miramonte Drive
Moraga, CA 94556
USA

We thank our partners for their continuing financial support

Cystinosis Foundation New Jersey Chapter
Orphan-Europe
Raptor Pharmaceutical Corp.
Sigma-Tau Pharmaceuticals
Tromp Beheer BV

When moving, please remember to notify the Cystinosis Foundation of your new address.