Cystaran Provides Ophthalmic Delivery of Cysteamine

By Karen Kuphal PhD

Providing an ophthalmic delivery of cysteamine - CYSTARAN™ (cysteamine ophthalmic solution) 0.44% is approved by the U.S. Food & Drug Administration (FDA).

The need for managing debilitating ocular manifestations of cystinosis has become increasingly important. One such ocular manifestation includes corneal cystine crystals, which are known to progressively worsen with time and do not spontaneously resolve. It is these ocular cystine crystals that are considered partly responsible for clinical symptoms of photophobia, recurrent corneal erosions, and secondary blepharospasm, which can complicate longstanding cystinosis¹.

Despite the benefits of reducing cystine levels systemically, cysteamine that is taken orally is not effective in reducing corneal crystals or the accompanying secondary symptoms of photophobia, blepharospasm, and eye pain²,³,⁴. This is due to the lack of blood supply to the corneal stromal cells, thus limiting availability of oral cysteamine to the eye. Acknowledging the need to investigate an ophthalmic route of delivery, a formal clinical trial of all the complications associated with this rare disease. Since then, we have had much experience and so it is with pleasure that we went to this conference.

This year, we stayed for two days in Paris, staying on site in the very comfort-

Support Group Launches in Brazil

By Xenia and John Cesar Mota

The Cystinosis Support Group - Brazil is the only organization of its kind in Brazil. Established one year ago, the group is not-for-profit and its members are individuals and families who are affected by cystinosis. We serve our

[See BRAZIL page 6]
From the President’s Desk

Dear Friends,

This has been an eventful year in many respects and for some of our friends here in the United States, lives have been disrupted over the past few weeks since Hurricane Sandy wreaked its havoc. To everyone who endured the super storm of the century, our hats are off to you. We hope those who sustained any type of loss may recover quickly. Please know our thoughts are with you all.

Thirty years ago cystinosis was a secret and an abysmal few physicians were familiar with this rare disease. Today cystinosis is no longer a secret. It is literally known around the world. Since expanding our mission internationally 15 years ago, the Cystinosis Foundation has assisted others in the establishment of support groups within their country and is pleased that 15 nations currently have such support. This includes Australia and New Zealand, Belgium, Brazil, France, Germany, India, Iran, Italy, Mexico, The Netherlands, South Africa, Spain, United Kingdom and Venezuela.

We encourage you to visit the Rare Connect web site at www.rareconnect.org, an online forum to connect with other individuals coping with cystinosis around the world. This site is available in five languages, English, German, French, Italian and Spanish, and offers translation services. An excellent resource for reliable information, the Cystinosis Foundation was instrumental in bringing together our international community on RareConnect.org. You may learn more about cystinosis, post your own thoughts and interact with others, as well as have your comments translated into the other four languages.

2012 marks FDA approval of Cystaran, an ophthalmic cysteamine treatment for cystinosis in the eyes. We began on this path together in 1986 and are eternally grateful for the commitment of William A. Gahl, MD, Ph.D., at the National Institutes of Health and many others over the past 26 years who never once gave up. Thank you to Sigma-Tau Pharmaceuticals, for its unswerving dedication to the cystinosis community.

The Cystinosis Foundation, in partnership with the Cystinosis Research Foundation, launched the Cure Cystinosis International Registry (CCIR) in 2010 and currently 34 nations are represented in this registry dedicated solely to cystinosis patients. This would not have been accomplished but for the strength of our international community working together. If you have not already done so, we encourage you to register today at www.cystinosisregistry.org. Betty Cabrera, MPH, curator of CCIR, is available to assist and answer any questions.

We hosted a successful 7th International Cystinosis Congress in France this year for 265 guests, our highest attendance at any conference to date. This well regarded educational program is expanding to Latin America in the spring of 2013 and to the East Mediterranean as well in our efforts to reach individuals and families affected by this ultra rare disease.

Early on I perceived that there is something exceedingly special about individuals coping with cystinosis and have yet to be able to describe my perceptions other than to say that in every instance, I have found they are very, very special individuals, with extraordinary spirits. Perhaps Stephen Hawking, the greatest theoretical scientist of our time, put it best when he said: "I am one of the most lucky people on Earth. A disability I might never have expected to overcome, and an abysmal few physicians were familiar with this rare disease..."

Stephen Hawking, the greatest theoretical scientist of our time, put it best when he said: "I am one of the most lucky people on Earth. A disability I might never have expected to overcome, and an abysmal few physicians were familiar with this rare disease..."

The Cystinosis Foundation Newsletter is intended to report items of interest with regard to cystinosis. The Corporation neither promotes nor recommends any therapy, treatment, etc. The relevance of information printed in this newsletter should be discussed by the patient or family with their own physicians.

The editor reserves the right to make corrections as appropriate and in accordance with established editorial practice in material submitted for publication. Individuals and organizations referred to do not necessarily endorse this publication or the editor. Our hope is that this method of continued information will promote communication between patients, families and professionals and foster support among affected families.

The Cystinosis Foundation Inc. has been designated a non-profit charitable organization by the Internal Revenue Service under Section 501 (C)(3) of the Internal Revenue Code. The federal tax ID is: 94-2927892

The Cystinosis Foundation Newsletter is published by the Cystinosis Foundation, Inc. (888) 631-1588 or (925) 631-1588

Cystinosis Foundation President and Founder - Jean Hobbs-Hotz
Newsletter Editor - Valerie Hotz
Newsletter Printer - Diablo Digital Printing

[See PRESIDENT page 4]
Patient-Reported Outcome and Health-Related Quality of Life in Adults with Cystinosis: A Study Utilizing the NIH “PROMIS®”

By Angela O. Ballantyne, Ph.D. & Doris A. Trauner, M.D.
University of California, San Diego

Overview
Nephropathic cystinosis is a rare recessive genetic disorder in which cystine, an amino acid, builds up in the lysosomes of cells. The result of this lysosomal storage disorder is the accumulation of cystine crystals in organs throughout the body, leading to multi-organ dysfunction. Cystinosis is a fatal disease, and there is no cure at this time. The advent of renal transplantation and cysteamine therapy have served to extend the lifespan of individuals with cystinosis into adulthood, but they do not prevent the ultimate progression of the disease. For this reason, it is important to learn about the longer-term effects of cystinosis on quality of life in adults with the disease, as well as the final illnesses and causes of death in this population. This knowledge can then be used in the planning of and/or justification for emerging therapies and treatments for adults living with cystinosis.

Specific Aims
Aim #1: The first aim of the proposed study is to provide vital information on patient-reported outcome and quality of life in the expanding population of adults living with cystinosis. This will be accomplished using a well-standardized and psychometrically sound measure of outcome in chronic conditions, which

Michael Vellard, Ph.D. Serves as Scientific Advisor

The Cystinosis Foundation is pleased to announce Michael Vellard, Ph.D., Head of Lysosomal Biology at BioMarin, to serve as Scientific Advisor for the 1st International Cystinosis Congress-Beyond Borders in Latin America. At BioMarin Dr. Vellard directs research focused on finding treatments for rare genetic diseases, including MPS1, MPS6, MPS4A and Duchenne Muscular Dystrophy.

In 1992 Dr. Vellard received his Ph.D. summa cum laude in Microbiology from the Pasteur and the Curie Institutes in Paris, France, on the regulation of the transcription of the protoncogene c-myb. Dr. Vellard worked on Cystinosis during his post-doc training at the University of California, Los Angeles School of Medicine. His work in Genetics and Molecular Biology has been recognized by several prestigious awards and several patents.

Testimonials

Dear Cystinosis Foundation,
This summer was my first time at the International Cystinosis Congress and I enjoyed the time very much. The Congress was a pleasant experience for me. In my opinion an International Cystinosis Congress is an excellent chance to connect with other people who have the same disease. As a summary I would say: New people, new experience and I wish to thank you for the nice time we had together.

– Jana Bielau
Germany

Dear Cystinosis Foundation,
I found the conference really beneficial for myself. Not only was it really informative to find out about the latest research developments, it was also a particularly rewarding experience to meet other adults with cystinosis from all over the world. I was sad when the time came to say goodbye to everyone when it was finished. Wherever it takes place, I will definitely be attending the next international conference in 2014.

– Ami Froehlich
Board of Directors
Cystinosis Support Group United Kingdom

[See TESTIMONIALS page 4]
34 Countries Represented in Cure Cystinosis International Registry

By Betty Cabrera, MPH

There is no doubt that technology is revolutionizing the way that disease information is collected and shared through online research tools such as the Cure Cystinosis International Registry (CCIR). Thanks to the far-reach of the Internet, CCIR provides opportunities to cystinosis patients all over the world to participate in this hallmark project to accelerate the pace of medical progress for this rare disease. Three hundred eighty individuals from 34 different countries have seized the opportunity thus far. One cannot help but ask, where would CCIR be today without the incredible patients, families and doctors who believe and support this mission? As Curator of the registry, I have a unique vantage point from which to observe the commitment that people throughout the globe have to improving the lives of those with cystinosis.

The Curator is on the receiving end of all medical surveys submitted confidentially to the registry and is alerted whenever a new survey is completed. It is exhilarating to witness when consciousness of cystinosis and the registry is high. On any given day persons in Brazil, Australia, Mexico and France may be simultaneously dedicating 30 to 45 minutes of their precious time to complete a CCIR survey. Portuguese, Spanish and French speakers may complete the survey in their native language, and soon Dutch speakers will be added to this list. In cases where access to a computer is limited or where there are other language barriers, registration assistance from a health care provider may be necessary. CCIR has come across many outstanding medical professionals who are committed to ensuring that the cystinosis patients they see receive the best care possible and are included in important projects like CCIR. They help spread the word about CCIR at conferences across the globe, recently in Belgium and China; they help recruit patients in hard-to-reach countries such as Egypt and Russia; and they selflessly volunteer to translate survey materials or find others who are willing to do so. And of course there are the organizations like the Cystinosis Foundation that help us immensely with making connections with patients and clinicians.

The cystinosis community is coming together like never before and CCIR is grateful to be a part of this. We invite those to who are members of the registry to track this progress by visiting the CCIR website and viewing the new map feature that allows one to see the areas of the world that our registrants come from. For those who have not yet registered, I would like to leave you with one thought that the American poet Ralph Waldo Emerson (1803-1882) penned, “Progress is the activity of today and the assurance of tomorrow.” Please find the time today to enroll in CCIR so that present and future generations affected by cystinosis will know a better tomorrow. We are here to answer questions about the registry and to help with registration. Please do not hesitate to contact us: www.cystinosisregistry.org or email curator@cystinosisregistry.org.
Creating Community in Spain

By Lilli Sanz

We are Javi and Lilli and Marta is our eight-year-old daughter.

She is a happy and funny girl, who is very sensitive, responsible, mature, self-sufficient and cuddly. She is just like every eight-year-old child, oh, and she has Cystinosis. Marta was diagnosed when she was a 6 months old, at Sant Joan de Déu Hospital in Barcelona. She has been taking Cystagon since she was very young. Medication has long been a part of her life, and she accepts it. Marta takes her medications with many questions, a measure of responsibility, but sometimes with anger.

When the doctor told us that Marta had cystinosis and apprised us of the consequences this disease could bring, our world came apart, as most likely it may have for many of you. But soon we found the Cystinosis Foundation website. In Spain there was no place where we could go and no one to answer our questions. Thanks to the Cystinosis Foundation, we were able to learn more and above all, we could send you an e-mail to the “support forum.” We are very thankful for how soon we received an answer. The reply from Anne Claire in France, who, besides, speaking Spanish very well, was really helpful and filled us with optimism, just like the mail from Seandradh in Ireland, who gave us strength to keep going. She is an example of courage. We will always be grateful for your immediate support at those moments.

We felt the need to have a support group in Spain since the very beginning. We did not want to make the path all by ourselves. In 2004, within the 3rd International Cystinosis Foundation Congress in Tarragona, we began our journey with the help of AIRG-España. It was the first time that some Spanish families gathered together and it was also the first time that they met families and patients from other countries. Dr. Roser Torra, president of the AIRG scientific committee opened her arms and embraced our efforts on this journey. Since then, we have been an active part of the association and we have been following the Cystinosis Foundation in every event all over Europe. For all this help, thanks to AIRG, to Dr. Roser Torra and to Carmen Caballero, for creating a support group about Cystinosis and for offering us all their support and help.

Also, we will never forget Dr. Rodés, from Instituto de Bioquímica Clínica in Barcelona, for her interest and support over the years. We were very lucky to count on the medical team of Hospital Vall d’Hebron in Barcelona.

Nowadays there are 58 patients in Spain, at all ages, some of them just a few months old to 47 years of age! Each year, new patients are diagnosed. Fortunately, being our disease a minority one, it is becoming more well-known and early identified. We also know the advances on Cystinosis treatment, and we trust that these will soon enhance our quality of life.

With the aid of Orphan Europe we are able to reach out to patients and families from all over Spain, contacting with hospitals and doctors who treat patients with Cystinosis. Step by step, we are able to reach many families, but not all of them. We still have a long road ahead.

Having the opportunity to talk to a family in our same situation and share the experiences makes things less difficult for everyone and we believe this is important. “Feedback” between families is comforting. Our main task is to support and give information. If the occasion permits it – AIRG-Spain annual meetings in Barcelona are very helpful. At these meetings we meet the new members of this community, share our stories with them, show them that our children can be as happy as any other child, that they have to have a normal life; go outside, play and enjoy life with their friends, fall and get up as much as it is necessary.

Jean, and later Valerie have always been close to us, giving us their support and sending us information. Thanks very much to the Cystinosis community all over the world.
EXPERIENCE from page 1

able Marriott Hotel. We live in the province and we arrived on Friday morning, just in time for our older children to visit Disneyland. We were able to attend the expert presentations, which we followed with great interest, while our youngest children were welcomed at child care and were well cared for there. Every presentation helps us to better understand the workings of this disease. Hearing directly from scientists involved in researching this disease was exciting for all of us.

And then there’s the physician who cares for our children involved in this conference. It is always impressive to hear as well, before all these people, and we are thoroughly impressed that this man who cares for our children is so well known — although of course we know him well.

The presentations that we enjoyed the most are, of course, those researchers who explain the advances in research and experiments that are underway to provide better care. What is also impressive is the involvement of all these people, doctors, nurses, researchers, parents and friends working to improve care for all patients with cystinosis.

Both of our elder children participate in the clinical trials and Raptor Pharmaceuticals has already greatly improved our lives! We hope that this treatment will be available for all very soon, even the new drops to treat cystinosis in the eyes. Learning all this new information at this conference is very important to us, as parents of three children with cystinosis.

And it is also true that I preferred to chat with other parents rather than listen about problems that are not yet ours. In this way, we are learning to protect ourselves a little. Fatigue wins the second day and our concentration is not as alert.

There are other aspects of the conference that are important to us, such as these moments where we can meet other parents and other families. While we talk, we share our experiences, our joys and we also share our difficulties. Our children made friends with other children who have the same problems, for example, learning that others sometimes do not take all the medications properly as prescribed.

BRAZIL from page 1

community by exchanging information and experiences, providing referrals to families, and working to get government approval of Cystagon so that our children may receive treatment. The Support Group performs other initiatives such as organizing events, supporting families in hospitals, providing information to people about lawsuits, as well as providing information about cystinosis to medical professionals. We develop partnerships with rare genetic disease organizations in Brazil and other cystinosis associations around the world.

During the first year of operation, the Support Group participated in organizing a regional conference on rare genetic diseases, which featured a presentation by an expert in cystinosis, participation in the organization and execution of a public demonstration in Fortaleza on Rare Disease Day, and participation in the 7th International Cystinosis Congress in France, sponsored by the Cystinosis Foundation. We have created a social network on Facebook, Grupo de Suporte à Cistinose Brasil.

As a result of our educational presentation before pediatric nephrologists at Albert Sabin Hospital in Fortaleza, four additional children have been diagnosed with cystinosis. Currently we are working with the National Health Surveillance Agency (ANVISA), hospitals and the pharmaceutical representative with the goal of achieving CYSTAGON regularization in Brazil and also to achieve covered care of cystinosis patients in Brazil. This is the work we must accomplish in order for our children to receive Cystagon treatment.

Finally, we are working with the Cystinosis Foundation to organize and participate in the first International Cystinosis Congress in Latin America, in association with XVI Pediatric Nephrology Brazilian Congress. This important meeting takes place April 30, 2013, in the city of Campinas in Brazil. For more information and to join our support group, please send us an email at gscistinose.ne.brasil@gmail.com. You are very important to us.

"Concentrate on things your disability doesn't prevent you doing well and don't regret the things it interferes with. Don't be disabled in spirit, as well as physically."

— Stephen Hawking
Giovanni DiMilia (USA).

Jess G. Thoene, MD, recipient of the 2012 Hobbs
Humanitarian Award, for his work developing the only
treatment for cystinosis and his service championing
rare disease. Dr. Thoene is director of the Biochemical
Genetics Laboratory at the University of Michigan.

Back Row, left to right: Merle Mund, Serena Scott, Ami Froehlich Front Row Seated: Sue
Scott, Maria Pekli, Deb Woodward, Sue Goulsbra, Kayla Drury and Thomas Goulsbra.

Gail Daniels (South Africa), Marjolein Bos (Netherlands) and Lesli King (USA),
Sigma-Tau Pharmaceuticals, enjoy the evening’s festivities.

Valerie Hotz, executive director of the Cystinosis
Foundation, Shelby Roach and Gary Roach (USA)
keeping it real at the congress.
Rezan Topaloglu, MD, Ann Marie O'Dowd, Neveen Sollman, MD, Sue Maguire, Gillian O'Rourke, William A. Gahl, MD, Ph.D. at the 7th International Cystinosis Congress, France.

Marjolein Bos (Netherlands) and Claudia Sproedt (Germany) share a laugh.

Left to Right: Holly Paine (U.K.), Danielle Daniels (South Africa), and Ami Froehlich (U.K.) enjoy a moment together.

Elena Levtchenko, MD., Ph.D., delivers presentation on growth in patients with cystinosis.

Alexandre Couppey (France), Yago Mota (Brazil) and Romain Couppey (France) became fast friends.

Guests participate in a workshop.
Guests participate in a workshop. Elena Levtchenko, MD., Ph.D., delivers presentation on growth in patients with cystinosis. Our thanks to Patrick Niaudet, MD, for serving as scientific advisor and presenting on renal transplantation in cystinosis.

Alexandre Couppey (France), Yago Mota (Brazil) and Romain Couppey became fast friends. Jana Bielau, Christian Sproedt, Anika Kuchta and Serena Scott.

Guests enjoy the warm summer evening at the conference, where simultaneous German and French translation was provided.
was developed as part of the NIH “Roadmap” initiative, and which meets the standards set forth in the FDA Guidance to Industry.

**Aim #2:** Although treatment is serving to extend the lifespan of patients with cystinosis, there is currently no available cure and the disease and/or its complications are fatal. A second aim of this study is to gather information on the final illnesses and specific causes of death in adults who had cystinosis and passed away during the past 10 years. Such data will serve to round out the information obtained from adults living with cystinosis.

**Participants**

We will recruit two groups of participants. One group will consist of at least 50 adults (ages 18 and older) diagnosed with cystinosis. (The goal is to obtain as many adults as possible, ideally more than 50.) This age group has been selected in order to examine outcome and quality of life in these individuals now that they are surviving well into adulthood. An additional subject group will be included as an ancillary part of this study. The second group of participants will be at least 20 parents (or nearest living relative) of individuals with cystinosis who passed away within the past 10 years. Information on the patients’ final illnesses and causes of death will be gathered from parents/relatives.

Thus, you are eligible to participate if:

(a) you are an adult over 18 years of age living with cystinosis,

OR

(b) you are a parent or nearest living relative of an adult who had cystinosis and passed away within the past 10 years.

**Procedures**

This is an online, questionnaire-based study that examines quality of life and outcome in adults with cystinosis. Participants will be asked to log in to a secure, online assessment center and complete a series of questionnaires that will take approximately 1 to 2½ hours. Questions will be asked regarding demographic background, health, medical issues, quality of life, and outcome in cystinosis. Participation in the study is strictly confidential.

**Measures**

**Measures for the Adult Living with Cystinosis:** This study will utilize the computerized online version of the PROMIS, which is the Patient-Reported Outcomes Measurement Information System. Data from all subjects will be combined, and participants will not be identified by name in any publication or teaching activities. Individuals from all areas of the U.S. and Canada, as well as from other countries, are eligible to participate.

**CYSTARAN**

began in 1986 by Dr. Kaiser-Kupfer at the National Institutes of Health (NIH) to assess the tolerability and efficacy of an ophthalmic cysteamine solution (administered as an eye drop) in patients already receiving orally administered cysteamine1. From 1986 onward, more studies on ophthalmic delivery were launched by researchers such as Dr. Gahl, Dr. Schneider, Dr. Thoene, and Dr. Tsilou. Through their collaborative efforts, these investigators delineated the risks, benefits, and stability (etc.) of several formulations of ophthalmic cysteamine1,2,3,5-7. Complemented by a partnership with Sigma-Tau Pharmaceuticals, a new drug application (NDA) for CYSTARAN™ (cysteamine ophthalmic solution) 0.44% was submitted to the U.S. Food & Drug Administration (FDA). In early October, FDA approved CYSTARAN™ for the treatment of patients suffering from corneal cystine crystal accumulation as a result of cystinosis. Dr. Gahl, the current Clinical Director of the National Human Genome Research Institute, summarized the approval “as an important advance for children and adults who suffer from cystinosis”. Since its inception, this project allowed an opportunity to demonstrate the fruitful collaboration among Sigma-Tau Pharmaceuticals, patient advocacy groups, academic institutions, and government agencies. This was further exemplified by Dr. Gahl’s statement that “FDA approval of this drug represents the culmination of a longstanding collaboration among the National Eye Institute, the Eunice Kennedy Shriver National Institute of Child Health and Human Development, the National Human Genome Research Institute and Sigma-Tau Pharmaceuticals. It also has involved invaluable cooperation from cystinosis advocacy groups.” Although approved, CYSTARAN™ is not yet commercially available to patients. Sigma-Tau Pharmaceuticals continues to work closely with the principal investigator, Dr. Bishop, in the ongoing clinical trial (Protocol 86-EL-0062) at the National Eye Institute (NEI) to transition availability of the product. Please look for updates regarding this transition on our company website www.sigmatau.com or www.cystaran.com. Questions may be directed to Lesli King, Sr. Manager of Patient Affairs, at 301-670-5450 or Lesli.King@sigmatau.com for any questions.

**Safety:** The most frequently reported ocular adverse reactions occurring in 10% of patients were sensitivity to light, redness, and eye pain/irritation, headache, and visual field defects.

7 Combined Analysis of Patients Treated with Oph- thalmic Cysteamine (CAPTOC); Clinical Study Report STP09294 on file at Sigma-Tau Pharmaceuticals, Inc.
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Thank you to all who have made a gift to the Cystinosis Foundation.
We are grateful for your financial support.
The Starfish Story
Original Story by: Loren Eisley

One day a man was walking along the beach when he noticed a boy picking something up and gently throwing it into the ocean.

Approaching the boy, he asked, “What are you doing?”

The youth replied, “Throwing starfish back into the ocean. The surf is up and the tide is going out. If I don’t throw them back, they’ll die.”

“Son,” the man said, “don’t you realize there are miles and miles of beach and hundreds of starfish? You can’t make a difference!”

After listening politely, the boy bent down, picked up another starfish, and threw it back into the surf. Then, smiling at the man, he said…

“I made a difference for that one.”
Making a Difference as Agents of Social Change

By Valerie Hotz

Since 1983 the Cystinosis Foundation has been an instrument of social change, working to empower families and patients by helping to create 15 support groups around the world, delivering services to areas where previously people were remained frightened and alone. We invite you to join us in our noble mission.

Recognized as a leader in increasing awareness about rare diseases, the Cystinosis Foundation has been a member of the National Organization for Rare Disorders since its inception and is a member of EURORDIS as well. Our international mission to serve individuals and families affected by cystinosis and to educate medical professionals helps people build meaningful lifelong connections and friendships, removing painful feelings of isolation.

You won’t see our results in the headlines, but everyday we are helping individuals and families affected by this rare metabolic disease throughout the world. Your financial contribution not only helps families, it also strengthens the scientific network that exists among cystinosis researchers, thereby benefiting patients in an additional way. We would not be where we are today without your support and we are eternally grateful for the financial support of thousands of people over the past three decades.

Thankfully, we live in a culture of innovation and a regulatory environment that encourages development of improved treatments, and yet we always seek out one-on-one social interaction to help each other in fundamental ways on our respective journeys with cystinosis. Ultimately, it comes down to the individual to make a difference and the members of the Cystinosis Foundation continue to make a difference every day. Nobody need wait a single moment before starting to improve the world. We hope you will join us in this endeavor.

Every dollar helps us accomplish our mission. Our work is not possible without the generous financial support of truly caring individuals and companies. Please consider making a gift to the Cystinosis Foundation today. You may make your contribution online at our web site www.cystinosisfoundation.org (click on DONATE) via secure PayPal processing, Visa, MasterCard, American Express or Discover, or by sending your check in the attached remit envelope.

The Cystinosis Foundation is a 501 (C) (3) nonprofit charitable organization. Your donation is fully tax deductible. Our federal tax ID is 94-2927892.

Who Knew?

William A. Gahl, MD, Ph.D. has a passion for playing softball in his spare time.
Deanna Lynn Potts Scholarship 2013

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 ______________

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Birth Date________________________ Gender(____) M (____) F

Social Security No.________________________________________

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Principle _____________________________________________________
Guidance Counselor _____________________________________________

Date of High School Graduation _____________________ Cum. GPA ___________

Name of College/University/Vocational School you will attend in the fall of 2011:

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: ______________

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 be on a family emotionally, socially, and financially. Children with cystinosis are living longer thanks to medical science and therefore embarking on careers that require education. Due to the financially draining medical costs it might prove difficult to send a child to college. We do not want to deprive our children of education in today’s world. Through this fund we hope to help some students.

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

APPLICATION PROCEDURE:
Applicant must submit by April 12, 2013:

Documentation 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? Why? The essay should be typed and double-spaced.

For complete guidelines for submitting an application, visit our web site at www.cystinosisfoundation.org

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, to encourage and support research for improved treatments and a cure and to mentor the establishment of support groups in other nations.

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.
New Patient Support Program Available in U.S.

By Valerie Hotz

Raptor Therapeutics, developer of delayed release cysteamine commonly referred to as RP 103 unveils RaptorCares, a new cystinosis patient support program that is currently available in the United States. This program provides tools, support and information to help patients and their caregivers manage cystinosis.

“The specific resources available include information about cystinosis that may be shared with the patient’s teachers, schools and childcare providers, management materials for adult and adolescent patients and an informational email series for parents who are learning how to manage cystinosis for their family,” says spokesperson Clair Johnstone. In addition, a helpful emergency room protocol and information sheet that may be shared with physicians and nurses in the emergency room is also provided. While RaptorCares is available now only for individuals within the United States, plans are in the works to make these resources available for families outside the U.S. next year. “All information provided will remain confidential, as patient privacy is very important to Raptor,” adds Johnstone.

Enrollees in RaptorCares will receive the most up-to-date information from the company on the progress of its application to the Food and Drug Administration for approval of delayed release cysteamine, which requires dosing every 12 hours. A copy of the 2011 global cystinosis survey conducted by Raptor Therapeutics, which reflects the triumphs and challenges of those living with this ultra rare disease, will be sent to every participant, as well as a free cooler for use while away from home.

For more information and to enroll in RaptorCares, visit the website at www.RaptorCares.com or call 1-855-888-4004.

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)

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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

You may make a gift to the Cystinosis Foundation online at www.cystinosisfoundation.org.
We thank our partners for their continuing generous financial support of our mission:

AIRG-France
Cystinosis Foundation New Jersey Chapter
Orphan-Europe
Raptor Pharmaceutical Corp.
Sigma-Tau Pharmaceuticals, Inc.

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