Families and Experts to Gather at 6th International Cystinosis Congress

Enthusiasm has been growing throughout our community around the world for the 6th International Cystinosis Congress. Over 22 physicians and leading medical experts will deliver presentations on the latest research at the International Cystinosis Congress in Lignano Sabbia- doro, Italy this September 23-26. Francesco Emma, M.D., Head of the Division of Pediatric Nephrology and Director of Nephrology, Bambino Gesu Children’s Hospital and Research Institute, Rome, serves as conference chair. Mara Mazzina, of Associazione Cistinosi Onlus, and Dr. Emma selected this year’s location. They have generously donated hours of their time to organize an excellent conference for families and scientists.

The site of this congress is beautiful Lignano Sabbia- doro, a popular vacation destination featuring the backdrop of a centuries-old pine forest and the Adriatic Sea. In a departure from previous conferences which have been hosted in hotels -typically in urban areas - the Village Ge.Tur is utilized as an athletic training facility, complete with dormitory style living and cafeteria. An art exhibit of works by cystinosis patients is planned to coincide with registration on Thursday, September 23 and a special salute to the contributions made by Dr. Jerry Schneider will be featured.

The Village is 1 ½ hours from Venice. Guests arriving at either Trieste’s Ronchi Dei Legionaire airport or Venice’s Marco Polo airport will have a free shuttle service available for transportation to the conference location. This year’s conference offers a relaxing environment, away from the hustle and bustle of modern living. If you have a need to be in touch with folks back home, please bring along your mobile phone and charger. The Village features numerous sports fields along with four swimming pools and tennis courts for guests’ use. Bicycles are available for hire.

Associazione Cistinosi Onlus and Mara Mazzina

By Valerie Hotz

The Cystinosis Foundation and Associazione Cistinosi Onlus are co-hosting the 6th International Cystinosis Congress in Lignano Sabbia- doro, Italy, September 23-26, 2010. Mara Mazzina has been devoting a tremendous amount of time coordinating this conference, beginning with selecting an ideally situated venue in the middle of Europe, the Congress Village Ge.Tur in Lignano Sabbia- doro. This location features the beauty of a centuries old...
From the President’s Desk

It is with great excitement that we are looking forward to the 6th International Cystinosis Congress in Europe. These international conferences were founded in Bergamo, Italy in 2000 by the Cystinosis Foundation. Over the past ten years successful conferences have been hosted in France, Spain, Ireland, The Netherlands, and Turkey. We have reached beyond borders to bring families and medical professionals together in this battle against cystinosis. Old friendships are renewed and new friendships continue to develop. The children enjoy these conferences where they are able to play with their peers who are coping with similar issues in daily living.

We are honored to have Francesco Emma, M.D., Head of the Department of Pediatric Nephrology and Director of Nephrology Laboratory, Bambino Gesu Children’s Hospital and Research Institute, Rome, serving as conference chair and Mara Mazzina, Associazione Cistinosi Onlus serving as co-host.

Twenty international medical experts in the field of cystinosis research are scheduled to speak, including Corinne Antignac, M.D., Ph.D., Director of the Inserm laboratory, Hereditary Nephropathies and Kidney Development, Professor in the Department of Genetics, Necker Hospital, Paris, Jerry A. Schneider, M.D., Dean for Academic Affairs Emeritus, U.C. San Diego, William A. Gahl, M.D., Ph.D., Clinical Director, National Human Genome Research Institute, National Institutes of Health, Elena Levchenko, M.D., Ph.D., Professor, Department of Pediatric Nephrology, University Hospitals Leuven, William van’t Hoff, M.D., Head of Clinical Research Facility, Great Ormond Street Hospital, London, Stephanie Cherqui, Ph.D., Assistant Professor, The Scripps Research Institute and Ranjan Dohil, M.D., U.C. San Diego Medical Center.

If you are unable to attend and have a ques-
Long-awaited Cystinosis Eye Treatment To Be Reviewed by FDA

By Valerie Hotz

Sigma-Tau Pharmaceuticals announced on May 17th the Food and Drug Administration (FDA) has accepted the filing of the company’s new drug application for its cysteamine hydrochloride ophthalmic solution. This investigational therapy is for the treatment of corneal cystine crystals in patients with cystinosis. The FDA granted Priority Review status, a designation given to drugs that may provide major advances or a treatment where no adequate therapy exists. Currently there is no FDA approved treatment for corneal cystine crystal accumulation, a condition which can lead to changes in visual acuity and photophobia (sensitivity to light).

The new drug application for cysteamine hydrochloride ophthalmic solution includes data from clinical trials conducted at the National Institutes of Health (NIH) National Eye Institute. Results of the studies indicate that the administration of cysteamine hydrochloride eye drops may be effective in the prevention and treatment of corneal cystine crystals.

“Acceptance of the NDA marks an important milestone for both Sigma-Tau and the cystinosis community,” said Greg Lapointe, Chief Executive Officer of Sigma-Tau Pharmaceuticals. “If approved, this treatment has the potential to offer important benefits for patients – many of whom are children – whose care has been limited due to a lack of therapeutic options and access.”

In 1986 the first study to treat corneal cystine crystals began at the National Institutes of Health under the direction of William A. Gahl, M.D., Ph.D., lead investigator. “Over the course of the past 24 years many different individual studies took place involving approximately 250 individuals. We know that it works. One should say it has been a long journey. Sigma-Tau Pharmaceuticals made a major contribution to this effort. For 15 years Sigma-Tau provided the drug by contracting with companies so the drug could be made available for this study and the company invested millions of dollars in this three-way collaborative effort involving patients, government and industry,” says Dr. Gahl.

“I think the EU will follow the lead of the FDA on this. However, a pharmaceutical company in Europe, such as OrphanEurope or Swedish Orphan, must initiate the application for approval to the EMU (the FDA equivalent in Europe),” he adds.

Sigma-Tau Pharmaceuticals was approached by the National Organization for Rare Diseases (NORD) in 1995 with a request to bring a product to market. While Dr. Gahl’s study continues, a portion of clinical records from his study were submitted in support of Sigma-Tau’s recent new drug application to the FDA.

Sigma-Tau’s application includes a requested dosing of one drop in both eyes daily every waking hour.

1st International Cystinosis Congress in East Mediterranean

By Jessica Jondle

The First International Cystinosis Conference in the Eastern Mediterranean - held in Ankara, Turkey in October 2009 - was a resounding success. As the first meeting of what will be a bi-annual event, families from all over the Middle East gathered to hear about the latest research from the international medical community, learn about treatment options from regional physicians, and network with one another to form lasting relationships. Although most families were from Turkey, other countries were represented at the conference, including Iran, France, and the United States. Medical presentations were given in English with simultaneous Turkish and Arabic translation.

Over the course of three days, conference participants heard encouraging news from the Cystinosis research community, including Dr. Corinne Antignac, whose lab in France has successfully developed a mouse model with the CTNS gene mutation. Other presenters included Dr. Francesco Emma from Italy, Dr. Elena Levchenko from Belgium, Dr. Craig Langman from the United States, and Dr. Rezan Topaloglu of Turkey.

There are a number of challenges facing Turkish individuals with cystinosis, as well as families throughout the Middle East. The gathering served as an important reminder to families of the critical nature of regular Cystagon dosing and marriage outside of tight-knit family circles so the disease is not perpetuated. Access to appropriate medical treatment can be an obstacle in the more remote parts of the country, and has also become a cause of concern for Iraqi families due to the hard-
Raptor Pharmaceutical Corp. Advances Delayed Release Cysteamine in Clinical Trials

By Ted Daley

Raptor’s DR Cysteamine, an investigational, delayed-release (DR) oral formulation of cysteamine bitartrate, is being studied for the potential treatment of nephropathic cystinosis. Raptor’s DR formulation may require less frequent dosing and reduce gastrointestinal side effects.

Nine patients completed the Phase 2b study conducted at UCSD in 2009. Key findings released in 2009 included:

DR Cysteamine demonstrated improved tolerability and the potential to reduce total daily dosage and administration frequency compared to immediate-release cysteamine bitartrate.

Twice-daily DR Cysteamine may achieve the same pharmacodynamic result while using a daily dose 30% lower than immediate-release cysteamine bitartrate capsules administered four times daily.

No adverse events recorded during the clinical trial were determined by the principal investigator to be possibly or probably related to DR Cysteamine. Nine adverse events recorded in the clinical trial were determined to be possibly or probably related to immediate-release cysteamine bitartrate.

“Raptor extends its thanks to all who have participated in the studies that have brought us to this point and appreciates the effort made by both patients and families for all study participation,” says Ted Daley, President of Raptor Pharmaceutical Corp.

Patient enrollment is expected to begin in spring. This will be a multi-center study with 4 sites in the US and 5 sites in Europe. The current Phase 3 protocol calls for:

1 Screening visit
7 consecutive days of clinical visits
Approximately 3 weeks of home use of the new drug
3 additional consecutive days of clinical visits

There will be blood draws and other evaluations. Travel and accommodations for participating in the study will be provided at no cost to the patient. Daily living expenses for a family member or guardian

Scoliosis in Cystinosis

By Craig B. Langman, MD

Scoliosis, or an angular deformity of the spine, is a common bone condition in children. It is most often idiopathic, or without specific known cause. Scoliosis is accentuated during periods of rapid growth, such as in adolescence. There are tests that can be performed by a physician on routine examination of the child that may identify the condition, but sometimes, falsely so. Definitive diagnosis depends on demonstration of the angular deformity by X-ray study of the spine. Referral to a specialist in pediatric orthopedics is encouraged for proper care.

Cystinosis is not associated with scoliosis in the absence of some other problem in bone. Such problems may be the coexistence of the more common problem, such as idiopathic scoliosis, with the rarer disease, or the result of more specific bone problems. For example, an asymmetric vertebral crush fracture may produce scoliosis in the growing child. Such fractures may result from corticosteroid therapy, for example.

Medications are generally not felt to be causative in cases of scoliosis. However, there are isolated case reports of the concomitant use of recombinant human growth hormone and the “development” of scoliosis. A review of over 900 children treated for over 3600 patient-years with recombinant human growth hormone did not find any relation between that therapy and the development of scoliosis.

Kidney transplantation may have bone complications, since most children with a functioning transplant have some form of chronic kidney disease (CKD). CKD is known to have associated bone disease, including the development of scoliosis.

Since scoliosis is a common disease of children, and there is no firm evidence that medications contribute significantly to its presence, we must conclude that the use of recombinant human growth hormone
The Cystinosis Foundation salutes the outstanding contributions of Jerry A. Schneider, M.D., made over a 45-year career dedicated to research on metabolic disease, with a special emphasis on cystinosis. Dr. Schneider announced his retirement as Research Professor of Pediatrics and Dean for Academic Affairs Emeritus from the University of California, San Diego School of Medicine. He began studying cystinosis, believed to affect approximately 500 individuals in the United States and 2,000 worldwide, in 1965 in his capacity as Clinical Associate at the National Institutes of Health. He earned his M.D. from Northwestern University, with postgraduate training at Johns Hopkins Hospital, and the Centre de Genetique Moleculaire, Gif-sur-Yvette, France, in addition to his work at the National Institutes of Health. He spent two years at the Imperial Cancer Research Fund Laboratories in London, as a Guggenheim Fellow and as a Fogarty Senior Fellow.

Dr. Schneider joined the faculty at U.C. San Diego in 1970 and in 1976 he reported cysteamine removed cystine from cultured cystinotic skin fibroblasts and from leukocytes in a cystinosis patient. This breakthrough led to a treatment for cystinosis patients, with Ruth Fenstermacher being the first patient involved in the initial cysteamine study. “Dr. Schneider saved her life. We have the highest respect and regard for him,” says Robert Fenstermacher, Ruth’s father. “Ruth was able to live a full life, attend college, enjoy a career as a medical assistant and was happily married to Dean Heinzerling, who also has cystinosis. She really enjoyed her work in the medical clinic,” says Helen Fenstermach-
er, Ruth’s mother. The Fenstermacher’s had previously suffered the loss of their three older children from cystinosis. “Dr. Schneider has done marvelous work in the field and is a fantastic researcher. We wish him all the best in retirement,” she adds. Ruth passed away in 2004 and is well remembered for her loving kindness.

Krista Mund was also involved in early cysteamine studies. “Dr. Schneider has been a friend as well as a research doctor for our family for 38 years. He made the diagnosis on our daughter when very few doctors were familiar with cystinosis and he included Krista in the Vitamin C study, as well as the cysteamine study. She has continued this drug for 30 years and seen many great improvements in the method of transport,” says Merle Mund, mother of Krista Mund. “We owe much gratitude to him for all the research he has done in the field of cystinosis. Because of him, many children with cystinosis will grow up to lead normal lives,” adds Krista’s father, Bruce Mund.

Grant funding from the Cystinosis Foundation beginning in the mid-1980’s provided Dr. Schneider’s laboratory the necessary support resulting in 15 published research papers, most notably the report on a method to measure cysteamine in plasma, which was extremely important. In the Cystinosis Foundation Newsletter, Volume XI No.3, Winter, 1993 Dr. Schneider wrote “When cysteamine was first used it was feared the drug might be excreted in the urine and could potentially build up to dangerously high levels in cystinosis children who might have diminished renal function. The development of this method allowed us to prove that this was not the case and that cysteamine is rapidly cleared from the blood even in patients with renal failure. This also demonstrated the importance of giving the drug as close to every six hours as possible, since cysteamine could no longer be detected after six hours.”

“An additional study was a comparison of cysteamine and phosphocysteamine. This paper showed the two drugs were equivalent and this study could not have been done if we did not have the method available to measure cysteamine in the blood,” wrote Schneider. The efficacy of treating cystinosis with cysteamine from very early infancy was established by Dr. Schneider during this time. He and his colleagues found that children who are started at a very early age do extremely well in terms of both renal function and growth.

Recent work under the direction of Dr. Ranjan Dohil and funded with grant support from the Cystinosis Research Foundation has led to the development of slow-release cysteamine, requiring twice daily dosage. Raptor Pharmaceutical Corp. enters Phase 3 of the clinical trials for delayed-release cysteamine this summer.

“Jerry has always had a gifted ability to select really good people in various other medical specialties, such as neurology and ophthalmology, and encourage them to research cystinosis, which has greatly benefitted patients,” adds Hobbs-Hotz, who first met Jerry Schneider after her grandson, Joshua Hotz was diagnosed with cystinosis in 1982.

“I felt hopeless in the face of this diagnosis and Jerry held out hope about an experimental drug called Cystagon. I asked what could I do? He told me he always wished for a foundation to provide educational and emotional support for families and the seed for the Cystinosis Foundation was sown that day.”

The Cystinosis Foundation was formed in 1983 and incorporated as a 501 (c) (3) nonprofit organization in California in 1984. As a means of bringing families together and educating physicians, patients and families about this rare disease, conferences have been hosted in the United States, Europe and the East Mediterranean for over 22 years. The Cystinosis Foun-
[See SCHNEIDER page 6]
**TURKEY** from page 3

The Cystinosis Foundation wishes you many relaxing and happy days fishing in the beautiful Pacific Ocean.

**PRESIDENT** from page 2

 tion you would like a medical expert to answer, please e-mail your question to cystinosis@quickline.it.

New to the cystinosis community are pediatric nephrologists Mikhail Kagan, M.D. and Alexsei Tsygin, M.D. of Russia. They will participate in the conference and we welcome them to the cystinosis community. Drs. Kagan and Tsygin have diagnosed cystinosis in Russia, and are actively participating in speaking of cystinosis at medical meetings. We are deeply grateful to Rezan Topaloglu, M.D., Professor of Pediatrics and Pediatric Nephrologist at Hacettepe University Faculty of Medicine who stepped forward to host the 1st International Cystinosis Congress in East Mediterranean in 2009. This outstanding conference welcomed 55 patients and their families to Ankara. You can read more about this conference and details of the expansion of our mission to include hosting a bi-annual conference in the East Mediterranean on page 5. We are proud of the scientists who have devoted their professional lives to cystinosis research and have given countless hours of their personal time in addition. We owe a debt of gratitude to J.E. Seegmille, M.D., J.D. Shulman, M.D. and V.G. Wong, M.D., as well as Jerry A. Schneider, M.D., Corinne Antignac, M.D., Ph.D., William A. Gahl, M.D., Ph.D., Michel Broyer, M.D., William van’t Hoff, M.D. and so many others. We are grateful for the dedication of industry leaders in the field of rare disease who have long been strong and loyal partners and continue with us as we expand our mission. Our effective partnerships include Sigma-Tau Pharmaceuticals, Inc., Mylan, Orphan-Europe and our newest partner, Raptor Pharmaceuticals, Inc.

We are grateful and give thanks for our families who have endured and given. Many thanks to our volunteers who keep us moving forward and the individuals and companies who contribute funds that help us wage our fight. I am reminded of a quote from artist and writer Mary Anne Radmacher, “Courage doesn’t always roar. Sometimes courage is the little voice at the end of the day that says I will try again tomorrow.” Wherever you are, whatever language you speak, we are in this together. There is a place for everyone and everyone is needed.

Jean Hobbs Hotz, President, Cystinosis Foundation

**RAPTOR** from page 4

accompanying a minor will also be covered.

Eligible patients must be on a stable therapeutic dose of Cystagon®, be able to swallow Cystagon® capsules whole, not have received a kidney transplant and take all medications orally, not through a gastric tube. Both www.clinicaltrials.gov and the foundation websites will be updated as new study sites come on line.

An extension study to determine the safety of long-term administration of Cysteamine Bitartrate Delayed-Release Capsules is planned. Patients who complete this Phase 3 study will be offered the opportunity to be treated with Cysteamine Bitartrate Delayed-Release Capsules until they are approved by the FDA or until Raptor Therapeutics withdraws its application with the FDA (for whatever reason).

Please contact your doctor or check out www.clinicaltrials.gov for further information. Raptor may be reached by email: clinicaltrials@raptorpharma.com or phone: 1-888-270-3828.

**SCHNEIDER** from page 5

was added. The Cystinosis Foundation has assisted in the development of ten patient support groups in various countries. “Due to Jerry’s endeavors, the isolation once felt by families is no longer felt. His guiding hand was always available. Having known and worked with him has been a humbling experience,” says Hobbs-Hotz.

Thank you Dr. Schneider. The Cystinosis Foundation wishes you many relaxing and happy days fishing in the beautiful Pacific Ocean.
Danielle Daniels Story
By Gail Daniels
Danielle’s Mother

Danielle’s journey with Cystinosis is in many ways the same as others suffering from this devastating disease. She was diagnosed with Fanconi Syndrome at the age of 2 years, and only at 12 years of age was she diagnosed with Cystinosis.

The lack of knowledge, medication and support was the beginning of a struggle with daily challenges and uncertainty. Doctor’s appointments, hospitalization, leg operations and feelings of discomfort, nausea, tiredness, weakness, headaches, etc. have become a new normal. Yet, despite all of this, Danielle’s spirit of endurance, love for life, positive attitude, sense of humour and faith in Christ are remarkable qualities that makes her a joy and an inspiration to all who know her.

Danielle’s determination to live as normal a life as possible has earned her the First Principal’s Award, “The Ernst van Dyk Award for Enthusiasm” in grade 4. This was a wonderful acknowledgement, that despite her many hospital visits that caused absence, she would find out about the work she had missed and always completed all her tasks with excellence.

Following her love of horses, Danielle started riding as therapy in 2004. She enjoyed this very much and looked forward to spending time with them, cleaning, brushing, feeding and riding. She unfortunately had to stop horse riding as she had started swimming, which clashed with the horse riding times. She is a great swimmer and for ½ years competed in many swimming gala’s for the disabled. She competed against a South African Para-Olympic Gold medallist and others who had already gained Provincial colors. She did very well and showed promise and receiving an award for the most improved swimmer in 2006, as well as a trophy for a new record in 100m breast stroke. In 2007 she was asked to represent our province, but declined, saying that she enjoys swimming, not competing. She also tried playing guitar and piano, but decided she prefers listening and enjoying music.

As a committed Christian, Danielle has also been involved in many ministries at church. She manages the sound desk on Sunday evenings which involves setting up of all sound equipment and controlling the sound during the church service. She does the filing of the music sheets for the bands and enjoys being an assistant leader at Junior Youth.

Another of Danielle’s passions is visual art. She has art lessons once a week and enjoys painting African landscapes in acrylic on canvas. She finds great pleasure in mixing the colors and getting it “just right.” She also studies photography at Tygerberg Art School. This has become a great favorite of hers and she doesn’t go anywhere without her camera. She has a wonderful ability to capture the most marvellous scenes or moments through her lens. In June this year, she would like to select a few photographs, have them enlarged and framed and complete a number of paintings and put them all on auction in order to raise funds to attend the International Cystinosis Conference in Italy in September. She loves being busy and it is important to her to be a part of something.

It is wonderful seeing her happy and we have many people to thank for that, as they give of their time to contribute to her life. She is loved by so many and as a family we appreciate all who contributes to that. The nursing staff and Dr. Peter Nourse at Tygerberg Hospital are a wonderful encouragement and Danielle loves her monthly visits with them. They are interested in all she does and make her feel very special. Dr. Nourse is kind, but also very strict when he reminds her of the importance of taking her medication regularly. He has given her long talks on being responsible and not depending on Mom and Dad too much. Danielle will turn 17 this year and is consequently in the process of transferring to a nephrologist for adult patients. Dr. Nourse still sees her alternatively to help her in this transition.

One of the greatest things that happened last year, was the discovery of the Cystinosis Family Support Forum. Until then we had only had the advice from Dr. Nourse and information via the internet. Danielle has never met anyone with cystinosis and therefore this is often a lonely road for her to walk. Since joining the Support Forum, we feel part of the global cystinosis family. This is a tremendous privilege and encouragement, and for the first time we feel that we can speak to someone who really understands our situation. We are extremely grateful to everyone for their help, support and genuine concern.

Danielle has great dreams and aspirations and as parents we want to support her in all her endeavours as far as possible. Like many other Moms, I have also resigned from my profession to take care of her, making every day count and trying to live without regrets. I have found that living purposefully has contributed to Danielle’s quality of life in a way that it touches her spirit and demeanour which results in a happy and spiritually whole person. We are aware that the road ahead will have many challenges, but are confident in the hope and assurance we have in Jesus Christ, as we continue to trust Him, even when we don’t understand, for without His sustaining grace this journey would be unbearable.
Faces in the Crowd…

Daniels Family from Cape Town, South Africa. Front row, Danielle Daniels. Back row, Colin, Dane and Gail. Gail has established a patient support group in South Africa with the assistance of the Cystinosis Foundation.

Left to right: Lyndsey Beeler and Shannon Paju share a laugh at the Cystinosis Research Foundation Family Day, 2010.

Serena Scott
Cystinosis Support Group - Australia

By Susan Scott

A special greeting from Australia to all of our cystinosis families.

Our cystinosis support group in Australia is going well. We have accomplished quite a number of things since we last wrote to you. In 2006 we had our very first cystinosis conference in the southern hemisphere. Cystinosis families from all over Australia and also a family from New Zealand attended. With the wonderful help of Dr. Paul Roy, from Sydney, we had our two day conference in Perth Western Australia. Dr. Roy has been involved with cystinosis patients for many years and his experience and information was much very much appreciated by all that attended.

There are approximately 35 cystinosis patients in Australia. Because of the large distances between states there is not a lot of opportunity to meet in person. However, our families are in constant contact with each other and very supportive of new families of which we have had a few over the last year or so. There have also been a couple of successful kidney transplants. An article about one of our Australian cystinosis lads appeared in a popular women’s magazine. It was his 2nd kidney transplant.

In 2008 we had two families and one Australian doctor able to attend the international conference in Ireland. We are hoping to have two doctors and maybe five families looking at going to Italy this year for the conference. A great deal of fund raising is planned by families to get to the conference.

In January our Cystinosis Support Group was approached by an organization called SMILE to promote International Rare Disease Day, held 28th February this year.

SMILE is an Australian charity that helps families whose children suffer from rare diseases. We accepted this challenge and organized a Quiz / Auction night for 27th February.

From what I understand we are the only support group in Australia that is attempting to promote this important day. We have had a great deal of support from the surrounding businesses. They have donated products and vouchers to us. The newspapers and radio stations have been approached so we will hopefully have some good coverage of our event which also promotes cystinosis awareness.

Many people have enquired about the eye ointment that was presented by Dr Geoff Zhang at the conference in Ireland. A number of the Australian children have been using the ointment twice a day for over a year. Photographs were taken before they began using it, and then again a year later.

So far results have been good according to Dr Stephen Alexander, of Westmead Hospital. The ointment is quite thick and tends to blur vision after administering it. This can cause a problem for those that need to drive cars. The nighttime dose administered as the children go to bed is fine. One patient is using a combination of eye drops in the day and ointment at night. If anyone is wishing to learn more about the ointment please contact me and I can give you Dr. Alexander’s contact information. He is happy to speak with pharmacists about the ointment.

One of our newest cystinosis dads is involved with web page design and offered to do us a new Australian cystinosis web page. He and his team have done a wonderful job. We have had many comments from all over the world about the animation showing the function of the cells and how cystagon helps remove the cystine. We really appreciated all the time and effort it has taken to build the new site. If you would like to check it out you may visit our web site at: www.cystinosis.com.au.

So that brings us up to date with what is happening in Australia. We have been approached about getting families and doctors together in May in Melbourne Australia. A visiting doctor from the United Kingdom will be in Melbourne at this time so we are waiting on dates and times of this event.

We look forward to seeing many of our friends around the world this September in Italy.

Until then we wish you all a healthy 2010.
Dutch Cystinosis Group

By Marjolein Bos

It is a great pleasure for me to inform you of the latest news of our Dutch Cystinosis Group. We have a big advantage living in a small country. When we organize an annual meeting in the middle of the Netherlands, all members of our group are able to attend this meeting with a maximum of a 1 ½ hour car drive. Approximately 80% of all patients with cystinosis in our country are also member of our Cystinosis Group. The secret of this high percentage is the constant support of Professor Elena Levchenko, M.D. Dr. Levchenko always attends our meetings and provides reliable, updated medical information.

Last year in 2009 our annual meeting was in June and we focused this meeting on the psychological problems experienced by adolescents and young adults dealing with cystinosis. A specialized psychologist was invited to give information about self management and adherence to the therapy.

This year we will have our annual meeting on November the 6th. The items we will present are;
- News and update on specific cystinosis developments gathered at the 6th International Cystinosis Conference in Venice.
- The measurement of cystine in the leukocytes, including what patients and parents ought to know about this important test.
- From The Netherlands, we will attend the International Conference with a group of 15 persons of which 5 persons are young adults dealing with cystinosis. They hope to meet other young adults to share and learn from each other, and have a lot of fun together in the process!

This year we will use our Dutch experience with the Cystinosis Group, to start a special organization for the Belgium patients and their families. The first appointment for this activity is in April. I am hopeful that at the end of this year we will be able to send invitations for a Belgium annual meeting.

Cystinose-Selbsthilfe was Established in Germany 20 Years Ago

By Claudia Sproedt

It began in 1990 with 5 families and some creative ideas about how to build an information network to inform all families and doctors about cystinosis treatment. In Germany that was all new. A lot of people had no idea about cystinosis and even less what this meant to the families involved.

Thanks to the efforts of Professor Erik Harms and Dr. Georg Wolff we could start with a small group of “cystinosis families”. Today we represent 80 families in Germany, Austria and Switzerland. Our representatives have been and still are visiting international conferences all over the world to collect information and spread the word of news coming from Germany. As an example, the enteric coated capsules discussed throughout the world right [See GERMANY page 11]
MAZZINA from page 1

pine forest and the Adriatic Sea, combined with a modern conference facilities and accommodations for guests that include two outdoor swimming pools and (just in case the weather does not completely cooperate) two indoor swimming pools, as well as tennis courts, soccer field and basketball courts for energetic guests. A zoo, Parco Zoo Punta Verde, is located nearby.

You may visit the web site to view the site of this year’s congress by going to www.getur.com/Convegni_Congressi/Sfoglia_Brochure/Brochure_Convegni.html.

In addition to managing the Associazioni Cistinosi Onlus since it was established ten years ago, Mazzina also works full time at the Chiavenna Town Hall, often working on Saturdays during election periods. She has two children, Riccardo and Diana. “During my free time I dedicate myself to my family, my house and the Cystinosis Associazione,” says Mazzina.

Ricardo has cystinosis and is attending the second year of high school, specializing in science subjects. He enjoys doing sports, especially skiing in winter, swimming, and riding his bicycle. He also loves reading novels and playing PlayStation. Mazzina’s daughter, Diana, is 26 years of age and graduated in foreign languages. We are extremely grateful to Mara for her creativity and dedication to helping plan the upcoming conference and to Diana for devoting many hours translating numerous emails for us.

SCOLIOSIS from page 4

is unlikely as its cause. However, a prudent approach might be to discontinue, temporarily or permanently, the use of the growth product during the evaluation and follow-up of scoliosis, since rapid growth spurts may be associated with progression of scoliosis.

Dr. Craig Langman is The Isaac A. Abt, MD Professor of Kidney Diseases at the Feinberg School of Medicine, Northwestern University, Head of Kidney Diseases At Children’s Memorial Hospital, Chicago IL.

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Burwell RG 2003 Aetiology of idio-


GERMANY from page 10

now have been in use on a private trial and error base by German children for several years already, what finally helped pushing the idea of the new capsules that will be available soon. The new booklet Orphan-Europe printed in German as a parent’s information about cystinosis is based on a thesis written by Anika Kuchta, an 18 year-old member of our group.

To our members we offer annual meetings with medical and social support, a website and a national email support group. We are proud and happy to be part of the international community of cystinosis families. Some of us will be in Italy, others will be thinking of you.

Cystinosis Support Group in Germany sends best wishes.

Cystinosis Foundation Message Forum Has New Look

Please take a moment and check out the new Message Forum on the Cystinosis Foundation website at www.cystinosisfoundation.org. We are really excited about this new look and its easy navigation. We encourage you to join and contribute to discussion topics or create a new one that is of interest to you.

The Message Forum is a wonderful way to stay in touch with friends around the world and to make new connections as well. This is an excellent opportunity to share your story with other families and find new information on a variety of topics. www.cystinosisfoundation.org

Arts and Crafts Exhibit at Lignano Sabbiadoro Congress

The Cystinosis Foundation is pleased to sponsor an exhibition of the work of our patient community at the 6th International Cystinosis Congress. The rich artistic talent of many of our friends has come to our attention and in the spirit of celebrating these many diverse gifts, we are making arrangements to present a small exhibit.

If you are interested in participating by bringing a few of your pieces to display at the conference, please contact Valerie Hotz at v.hotz@att.net for specific details. Please include information about your art (painting, photography, ceramics, jewelry, needlepoint, etc.) and display needs. The deadline for submission is July 15, 2010.
I would like to take this opportunity to introduce myself. Although I have been a part of this amazing organization for many years, my involvement over the past 6 months has changed. The Cystinosis Foundation unexpectedly lost our dear friend and leader, Mr. Gaetano Di Benedetto, in August 2009. It is my honor to follow in his footsteps and continue his legacy. I am humbled by this new responsibility and hope to do the Cystinosis Foundation proud.

We were honored to have a very special guest with us that evening, Cystinosis California’s own, Ms. Valerie Hotz, who reported encouraging news about Dr. Stephanie Cherqui’s developments in gene therapy research. While recognizing there are many years of research ahead, Dr. Cherqui is optimistic about the potential for conducting clinical trials for stem cell therapy to treat cystinosis in the future. We are pleased Valerie was able to see first-hand what a special group of supporters the New Jersey chapter has and to share this wonderful annual event.

May God Bless all who cope with cystinosis, especially our very own, Dina Lotrecchiano. And may all of our beautiful memories of our beloved angel, Mickey Pugliese, be what inspires us to fight harder each and every year!!

With sincere appreciation,
Annie Basile
President, Cystinosis Foundation of New Jersey
Cystinosis Foundation UK Celebrates Tenth Anniversary

The Cystinosis Foundation UK has now been in existence for over 10 years. The first meeting of the founding committee, brought together by Jonathan Terry, was held on October 22nd 1998. Present were Gareth Jackson, who became chairperson, Paul Doyle, who agreed to be treasurer, and several members of Jonathan’s family. They drafted the constitution and made plans. The first newsletter went out in February 1999 and the foundation was registered as a charity with the Charities Commission during March. The official launch was at a meeting for members in St James’ Hospital (Jimmy’s) Leeds on April 22nd 1999. The foundation ended the year with £2,000 in the bank.

A national conference took place in Walsall in 2001 where Cystinosis expert Dr. William Van’t Hoff, of Great Ormond Street Hospital, agreed to become President. By the end of 2001 we had raised £25,000. This allowed the first research grant to be made in 2002 to a research group at Sunderland University, represented by Dr. Don Cairns, for an initial study of the possibility for a Prodrug approach to the delivery of Cystagon. Subsequently, Dr. Cairns moved to The Robert Gordons University in Aberdeen (where he has undertaken further cystinosis research), but the work has continued at Sunderland led by Dr. (now Professor) Roz Anderson. We continued our support for Sunderland University during 2003 with a research grant of £16,000 per year for 3 years.

In 2003 John Terry, Jonathan’s father, became a trustee and in 2004 the appeal to raise £100,000 for a research fund was launched. 2004 also saw a National Conference held at Biocity in Nottingham. 2005 was the year that Roy Forsyth became a trustee and Matt Blackham took over the website and by the end of that year the Research Fund Appeal stood at £61,000 and a small grant was made to Don Cairns to purchase a laboratory oven to assist in his research.

Paul Doyle announced at the trustees meeting in December 2006 that the £100,000 target had indeed been achieved, and we decided to then attempt to raise a further £100,000 since research can be very expensive. A request to fund a research assistant at Sunderland University for 1 year was granted. Also, under Paul’s guidance, Fundraising Standards Board approval was obtained in November 2007.

By July 2008 the Appeal Fund had reached £143,000 and during that year we received three further applications for funding, which committed most of the monies raised. It was agreed to support the work at Sunderland for 3 more years, to support work in Don Cairns group, at The Robert Gordons University, attempting to develop both a suppository delivery of Cystagon and an eye gel, and we also agreed to provide funds for an ‘Adult Cohort Study’, which aims to establish the medical problems met by adult cystinotics, led by Professor John Feehaly from Leicester and Dr. Van’t Hoff.

Our continued support of Professor Anderson and her team at Sunderland University is also beginning to yield results. They have shown that at least one of their prodrug1 formulations does work to release cystine from cystinotic cells and is non-toxic, both of which were key steps in the development. Successful development of a prodrug will greatly assist in the administration of Cysteamine1.

In November 2008 Paul Doyle retired from his position as treasurer and trustee. Despite having no family connection with cystinosis he has given nine years valuable service, providing sound guidance throughout. He was succeeded as treasurer by Gareth Jackson, our long serving chairperson. Meanwhile, Roy Forsyth assumed the position of chairperson and two new trustees, David Benford and Neil Sugden joined the committee.

1: What is a prodrug? A prodrug is a chemical compound which can carry a drug into the body in a form which gets it to at least past the stomach before it breaks down to release the drug. In the approach being followed at Sunderland University it is carried right into body cells before being released.

NORD 2010 Partners in Progress Gala

At the invitation of Roland Rutschmann, Vice President of Orphan-Europe, Valerie Hotz attended the annual National Organization for Rare Disorders (NORD) Partners in Progress Gala last month representing the Cystinosis Foundation. The gala was hosted at the Mellon Auditorium in Washington, D.C., an August and historical venue where, among other things, the North Atlantic Treaty was signed by President Harry S. Truman in 1949. It did not escape notice that vital and effective international partnerships were signed into law in the Mellon Auditorium over sixty years ago and today the spirit of cooperative venture continues to thrive through strong relationships between industry and the many rare disorder communities. The Cystinosis Foundation has been a national member organization of NORD for over 25 years.

Orphan-Europe and the Cystinosis Foundation have partnered over the past decade to bring excellent educational family conferences that include reports from medical professionals on the latest in cystinosis research and treatments. In addition to providing emotional support and better understanding to affected individuals and [See NORD page 16]
International Registry Launches July 2010

By Valerie Hotz

At the present time there is no registry existing in the United States for the cystinosis community, but all that will change in July when the Cure Cystinosis International Registry (CCIR) is launched by the Cystinosis Research Foundation. The Cystinosis Foundation endorses the CCIR and recognizes this registry as an invaluable tool in the improvement of care and the development of potential new treatments.

Working as a committed partner in this project, the Cystinosis Foundation will provide complete information about CCIR at the International Cystinosis Congress in September, as well as an update on the European registry by Patrick Niaudet, M.D. The Cure Cystinosis International Registry will collect basic information on individuals with cystinosis on a strictly volunteer basis. All personal information will be maintained in a secure database accessible only by a professional curator. Information that could identify participants and their family members will not be shared without express prior approval.

In the registry will allow physicians and researchers to identify trends, to design clinical trials for potential new therapies and to recognize the most effective treatments. Patient profile information will enable researchers to learn more about the scope and impact of cystinosis on individuals and their families. The Cure Cystinosis International Registry will be global in nature and will strive to achieve global registration of the cystinosis community.

“Recruitment for clinical trials can often take a long time especially for a rare disease like cystinosis. Participation in this registry will help speed up the recruitment process and help facilitate and expedite clinical trials. It is our hope the registry will lead to advances in care and treatment that will ultimately benefit those who suffer from cystinosis and their families. We believe participation in the registry will offer every member of our community a role in the quest for a cure for cystinosis,” says Nancy Stack, Trustee of the Cystinosis Research Foundation. Patients who register with CCIR will have the benefit of having access to information about new treatments and clinical trials and will be able to access information regarding clinical trials for which they may be eligible.

Leading cystinosis experts and members of the patient advocacy community will serve on the Advisory Board. For more information about the CCIR please visit the web site at www.cystinosisregistry.org. The Cystinosis Foundation endorses CCIR and encourages individuals and parents to consult with their physician and family and to participate in this important effort.

Annie Kwakkel, Dutch Artist and 2009 Recipient of Deanna Lynn Potts Scholarship

By Annie Kwakkel

My own physical body plays a big part in my art work and this is because of different experiences, such as dysfunctions, highly athletic performances - I was a driven gymnast for 10 years - and in general the necessity of our bodies. Without a body, you do not exist.

Because of these experiences, I am more aware of my own body, and the opportunities I have, because I know it is not obvious that everything works. I try to make people more aware of their bodies in a positive sense.

Everything I make begins with my fascination for the human body. Blood, flesh, faeces, organs, the solid matter what we are made of, this matter that makes us visible, makes us be. And about the void, between human bodies. Options to reach out, to make contact and to touch. This tension, this void, is most important. It stands for the possibilities, the options for the future. I work with intensely fragile material. Flower petals for instance. They are so fragile that often it almost is not there. And also transparent thread, glass, fabric and sheet. To be or not to be, the material and immaterial flow over and in each other.

Education is always costly, but at Art Academy besides paying for books, we also have to buy all our materials, tools and everything. I make a lot of movies, which requires a lot of camera equipment. We have regular excursions to interesting art places, such as Rome, Venice and New York. These excursions are also very expensive. The Deanna Lynn Potts Scholarship helped me out tremendously and made it possible for me to make an excursion to Venice in the autumn of 2009, which I enjoyed very much!

Editor’s Note: Annie Kwakkel is 23 years of age and graduates this month from the ArtEZ Academy CABK, located in Zwolle, The Netherlands.

Annie Kwakkel with one of her many art pieces.

She was awarded the Deanna Lynn Potts Scholarship by the Cystinosis Foundation and was able to pursue her creative passion. Annie will attend the 6th International Cystinosis Congress in Lignano [See KWAKKEL page 15]
In Loving Memory of Michelina Pugliese

May 2, 1975 – April 18, 2010

The Cystinosis Foundation is deeply saddened to hear about the passing of our very own Michelina Pugliese. Her story is truly remarkable and the memories of her will live on in us forever.

“Mickey” was born in New Jersey on May 2, 1975 to her loving parents, Domenico and Angelina Pugliese. About 9 months after she was born, the sweet, blonde haired, blue eyed baby spent most nights fighting high fevers, nausea and lacked an appetite. Immediately Domenico and Angelina went to doctors to find out what was wrong with their little girl. For more than two years she remained undiagnosed as having cystinosis, but was treated for various other illnesses. Gratefully, they were put in touch with a wonderful doctor, Joan Arboyd, M.D., who researched many avenues in order to find someone with a similar case as Mickey.

The doctor’s search went throughout the United States and led the Puglieses to a research institute in California that was treating these VERY FEW cases (at that time there were only 80 diagnosed cystinosis patients in the United States). In 1978, Mickey was diagnosed with Fanconi Syndrome and then shortly after that, diagnosed with cystinosis.

At that point, Mickey’s kidneys began to fail and her struggle became increasingly difficult. In addition to the kidney failure, Mickey began to lose her vision as well. She spent more than 12 years on dialysis and during that time became blind. Mickey became a part of a clinical study for a new drug called cysteamine, which today we know as Cystagon.

She was also a part of the clinical trial for the eye drops to stop the accumulation of cystine crystals in her eyes. Her medication schedule was grueling. Her mornings started with nearly a dozen different medications and her nights ended the very same way. Some medications were taken every 6 hours. She took them without a fight; she made it seem so easy. Due to her significant kidney damage, Mickey was at the top of the list to receive a kidney transplant. After two transplant failures, the Puglieses still remained hopeful that someday Mickey would successfully receive a transplant. On January 5, 1994, there was reason to celebrate. The third attempt at a kidney transplant was a success and thereafter Mickey and her family celebrated her “Kidney Day” every January 5th.

Things were copacetic for many years. Mickey had learned how to live comfortably with cystinosis, but she always remained hopeful that someday soon there would be a cure for the disease. In September 2009, things took a turn for the worse. Mickey was diagnosed with cancer. Just as she fought cystinosis for so many years, Mickey was determined to fight the cancer as well. After months of chemotherapy, her little body could take no more. On April 18th 2010, she became an angel, another angel to watch over those coping with cystinosis.

Her amazing spirit and zest for living was inspiring to so many. She touched the hearts of everyone she met and was always surrounded by people who loved her. We take comfort in all the beautiful memories we have now that she rests in peace.

Sabbiadoro, Italy this September and her art will be on display at a special exhibit of patients’ art work at the conference.

Cystinosis patients who are artists and are interested in sharing your painting, photography, ceramics, needlework or other crafts at the international congress this September, please send Valerie Hotz an email for more information v.hotz@att.net.

Prince Harry visits with Priscilla, a cystinosis patient at the St. Joseph’s Home for Chronically Ill Children in Cape Town, South Africa. See the full story in the next issue of the Cystinosis News.
The focus is on the common ground all people with rare disease share which includes the need for earlier diagnosis, more research, safe and effective treatments, for access to needed medications and for the development of a cure. Congratulations to EURODIS for drawing together the global rare disease community by establishing World Rare Disease Day.

Roscoe O. Brady, M.D. was the recipient of the Lifetime Achievement Award. Dr. Brady joined what is now the National Institute of Neurological Disorders and Stroke at the National Institutes of Health and remained there his entire career. He conducted pioneering research for over 50 years on hereditary metabolic storage diseases. His work has defined much of what is currently known about the biochemistry, enzymatic bases and metabolic defects of these diseases. Over the years Dr. Brady and his research team identified the underlying causes of Gaucher, Niemann-Pick, Fabry and Tay-Sachs diseases and developed methods to diagnose individuals with these conditions and detect carriers. They established the first effective treatment – enzyme replacement therapy – for Gaucher’s disease.

In 1968, Sami I. Said, M.D. undertook research at the Karolinska Institute in Sweden and successfully isolated a peptide that he believed existed in lung extracts. Dr. Said named the peptide “VIP” (vasoactive intestinal peptide) and ultimately established a line of research that he continues to research today. He and his colleagues have published more than 370 scientific articles on VIP and related topics, seeking to define the physiological role of such peptides, their relationship to disease, and their therapeutic potential. Dr. Said’s life’s work is now part of the basis for clinical trials underway at several European locations. VIP and other peptide-based therapies may have a possible application in the treatment of rare lung diseases.

John Crowley, President and CEO of Amicus Therapeutics, gave an impassioned speech about never giving up and shared the story of how he pursued a better treatment for Pompe disease, which affects his two youngest children. His strong commitment led him to leave his position at Bristol-Meyer Squibb, and become the CEO of Novazyme, a biotechnology startup that was conducting research on a new experimental treatment for Pompe disease. Novazyme was acquired by Genzyme, the world’s third largest biotechnology company and Crowley’s children received the enzyme replacement therapy for Pompe disease developed at Genzyme. Crowley credits the experimental trial with saving his children’s lives. Their story is told in the film, Extraordinary Measures, starring Harrison Ford and Brendan Fraser.

The Gala emcee was television and film star Patricia C. Richardson, (Home Improvement, Strong Medicine and The West Wing) who serves as the national spokesperson for Cure PSP: Foundation for PSP/CBD and Related Brain Diseases. Richardson’s late father, Lawrence Richardson, experienced PSP in the later years of his life. She served as emcee for the NORD Gala to help draw attention to the needs of all rare disease patients and their families.
and during the conference a competitive fencing match will be featured in the main gymnasium which guests are welcomed to attend.

Bring your swim suits - the weather won’t be a hindrance as there are both indoor and outdoor pools and you will need your swim suit to participate in Mack Maxwell’s water aerobics demonstration. It is for the entire family! Child care is available throughout both days and Sunday morning while parents are attending presentations. However, parents must be available to administer all medications to their child. Dialysis is conveniently available on site. Guests are required to provide in advance appropriate medical records to the medical supervisor at the Village Ge.Tur. For further details regarding dialysis, please email cystinosis@quickline.it for information.

Affordable, attractive rates are available at the Village Ge.Tur, with a single room rate of 58 Euros nightly, double room is 38 Euros nightly per person and family room (3-5 beds) is 36 Euros nightly per person. These rates include three meals daily and are available both preceding and after the conference ends, in case you would like to stay additional days. On Sunday afternoon, an optional trip to Venice is offered at 78 Euros per person. Please visit the online registration website at www.quickline.it. Click on “en” to change the page to English and go to “register” to complete your hotel reservation as well as to “download the submission form” to complete the full conference registration. If you have any questions, or wish to secure your booking by telephone, please call Quickline Agency in Italy at +39 040 773737.

The congress features the following three workshops: Workshop for Children with Parents Present with Dr. Francesco Emma, Workshop for Adolescents and Young Adults with Dr. William A. Gahl, and Dr. Marlies Ostermann and Workshop for Adult Patients with Dr. William Van’t Hoff and Dr. Marlies Ostermann. As always, there will be an informal poster session where families and patients can ask questions of physicians and representatives from industry. This conference is an opportunity to meet and connect with others facing the same daily challenges in a relaxing atmosphere conducive to conversation.

Highlights of exciting developments in cystinosis research will include a report from Dr. Dohil, U.C. San Diego and Children’s Hospital, San Diego, on long acting cysteamine. Results of Dr. Dohil’s study have given hope for the development of effective, better tolerated, twice-daily administered formulations of cysteamine bitartrate. This, in conjunction with the news from Raptor Pharmaceuticals that it is entering Phase 3 of clinical trials for its delayed-release cysteamine treatment at sites in Europe and the U.S. is very encouraging.

Dr. Patrick Niaudet, Hospital Necker-Enfants Malades, Paris, will update everyone in regards to “The new European registry” during his presentation. This is of special interest to families affected by cystinosis wherever they may live. There is an effort underway to establish a cystinosis registry in the United States and more information will be provided as it becomes available. The Cystinosis Foundation supports the formation of such a registry, which is completely confidential, and is of vital assistance in gathering details about this rare disease which will assist researchers in their progress towards better treatments and ultimately a cure benefitting all patients. Watch for specific details of the Cystinosis Registry in coming newsletters and on our website at www.cystinosisfoundation.org.

Corinne Antignac, M.D., Ph.D., Hospital Necker-Enfants Malades, Paris, focuses on “Genetics of cystinosis and prenatal counseling”. Dr. Antignac led groundbreaking research to identify the gene responsible for cystinosis in 1995, an important foundation paving the way to developments in gene therapy. Dr. Antignac will chair the professionals session on “New Frontiers”.

Stephanie Cherqui, Ph.D., Associate Professor at Scripps Research Institute will report on her exciting work. Dr. Cherqui is pursuing promising stem and gene therapy research with the goal of developing a cure for cystinosis and envisions the potential for clinical trials to begin in approximately 5 years.

Elena Levchenko, M.D., Ph.D., Universit"y Hospital Leuven, Belgium, delivers an update to families “on cysteamine treatment and adverse events” and Dr. Levchenko will chair the professional session entitled “Lessons from other diseases.”

Monte Del Monte, M.D., Professor of Ophthalmology and Visual Sciences and Professor of Pediatrics at the University of Michigan, will present on “Eye problems and the new cysteamine collyrium.” This topic is of special interest in light of Sigma-Tau Pharmaceuticals’ recent filing with the Food and Drug Administration in the U.S. of its new drug application for cysteamine hydrochloride ophthalmic solution, an investigational therapy for treatment of corneal cystine crystals in patients with cystinosis. The FDA granted Priority Review status, a designation given to drugs that may provide major advances or a treatment where no adequate therapy exists.

In 2006 Stephanie Cherqui, Ph.D., Associate Professor Scripps Research Institute began a project entitled, “Stem cells and gene therapy for cystinosis”. This study has demonstrated that transplantation of bone marrow stem cells or hematopoietic stem cells expressing a functional CTNS gene results in the abundant tissue engraftment of transplanted cells and decrease of cystine content (between 57-94% decrease). This treatment improves the eye, bone and kidney diseases of cystinosis. Dr. Cherqui is currently working on the ex vivo genetic modification of Ctns-/- hematopoietic stem cells to express a functional CTNS gene and build foundations for a future clinical trial for stem cell therapy to treat cystinosis. There is great optimism that clinical trials for gene therapy cystinosis treatment may be realized in 5 years time.

These are just a few of the many, many topics to be discussed. A professional session provides an opportunity for leading scientists to brainstorm topics pertinent to cystinosis research. Join us at the 6th International Cystinosis Congress to renew old friendships, share ideas, meet new people and make new friends. We look forward to seeing you in Lignano in September.

The Cystinosis Foundation gratefully acknowledges sponsorship support for this international conference from long-time industry partners Sigma-Tau Pharmaceuticals, Inc., Orphan-Europe, as well as Cystinosis Foundation, New Jersey and Associazione Cistinosi Onlus.
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 let-
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, USA.

(PLEASE PRINT)

Honor Circle 25,000 Yes, I want to become a member of the Cystinosis Foundation.
Patron 20,000 Enclosed are my membership dues of $______________
Lifetime 2,500
Visionary 1,000 No, I do not wish to become a member at this time, but I do wish to make a contribution.
Professional 200 Enclosed is my gift of $______________
Supporter 100
Family 50 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
When moving, please remember to notify the Cystinosis Foundation of your new address.

Cystinosis Foundation
58 Miramonte Drive
Moraga, CA 94556
USA

INTERNATIONAL CONGRESS
DUBLIN, IRELAND, 2008

INTERNATIONAL CONGRESS
BERGAMO, ITALY, 2000

INTERNATIONAL CONGRESS
NOORDWIJKERHOUT, NETHERLANDS, 2006

INTERNATIONAL CONGRESS
PARIS, FRANCE 2002

INTERNATIONAL CONGRESS
TARRAGONA, SPAIN, 2004

INTERNATIONAL CONGRESS
ANKARA, TURKEY, 2009

INTERNATIONAL CONGRESS
DUBLIN, IRELAND, 2008

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