



The Cystinosis Advocate

Volume 1, Issue 1

Spring/Summer 2008

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Twins with Cystinosis Receive Record-Breaking Kidney Transplants

The Gift of Life X 2

Cystinosis received national attention in December, 2007 thanks to Anjanelly (Anji) and Janelly (Nelly) Polanco. The identical twins with cystinosis each received a new kidney at the same time from the same donor in the nation's first simultaneous transplant for twins. The transplant took place at Children's Memorial Hospital in Chicago, IL.

The girls were ten years old at the time of their transplant. Both were on dialysis—Anji for one year and Nelly for for two and a half months. They were on the transplant waiting list for about six months. Their Nephrologist, Dr. Craig Langman, made a special request for them to receive kidneys from the same donor, so they could be transplanted at the same time.

The twins' mother, Rachel DaLomba, explained that all they were told was that the girls' kidneys came from a



Anji and Nelly Polanco

young victim of a car crash. The family has not yet made contact with the donor family. Rachel wanted to wait until the girls could fully understand the donor family's loss and what it means for them to give such a precious gift.

Cystinosis Diagnosis

Anji and Nelly were diagnosed with cystinosis in 1998 when they were eleven months old. Before being diagnosed, they were vomiting frequently and would not eat or gain weight. They were finally admitted to the Hasbro Children's Hospital in Rhode Island when they were ten months old, where they had tests run on them for a month.

Finally, one of the doctors, who Rachel remembers as a student or fellow, suggested they test for a disease she had read about in school—cystinosis. She was right. Once the twins' were diagnosed, they were quickly put on cystagon and other meds.

Challenges

Rachel DaLomba says vomiting has been one of the biggest challenges for the girls.

Continued on page 39



Miss United States 2007 Ashley Kazian Spreads the Word About Cystinosis

Ashley Kazian was crowned Miss United States 2007 in August, 2007. Since then, she has been on a whirlwind of public appearances and photo shoots.

Ashley is a friend of the McGinnis family. Twelve-year-old Laura McGinnis

has cystinosis. Ashley was named a spokesperson for the Cystinosis Research Network and tells others about cystinosis every chance she gets. She made an appearance May 3 at the 5K Run-Walk Fundraiser for Kacy Wyman in Bloomfield Hills,

Michigan and will be attending A Festival of Hope for Alexis and Evan LeBeau in Downer's Grove, Illinois on September 18.

Ashley took some time out of her busy schedule to update CRN on her experiences. See her letter and more photos on page 16.

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Message from the President

Christy Greeley



I am certain you will notice major changes in this edition of the Cystinosis Research Network's newsletter – beginning with a new title, *The Cystinosis Advocate*. The new design and title are meant to communicate and reflect CRN's mission and commitment to the cystinosis community. Over the last five years, CRN has grown to become one of the leading advocacy organizations for the cystinosis community, providing a complete source for family support, professional information, advocacy, and significant research grant provision in the U.S.A and rest of the world. We have embarked on a major program to extend our reach to a broader audience in organizational and geographic terms.

This newsletter is one of the principle vehicles via which we can communicate significant occurrences in the realm of cystinosis, focusing on capturing accomplishments and advancements the entire cystinosis community should be aware of. The objective is to provide comprehensive information where individuals can avail themselves of relevant information regardless of its source.

I trust you will find this edition of *The Cystinosis Advocate* insightful and relevant to you. In addition to the articles on research and advocacy, this edition captures incredible stories of courage and success from a variety of individuals. Our cover story details the groundbreaking double kidney transplant received by the 10-year-old Palanco twins last December. It is an amazing example of how far we have come and how a community can provide the necessary support to enable these individuals to prosper. You will also find encouraging stories of other individuals' life journeys and how they are successfully navigating life's challenges. Their stories are truly inspirational.

Overviews from our standing operating committees are included as well: Finance, Development, Research, Family Support, and Education and Awareness:

Finance - An Annual Donor Honor Roll has been instituted and will be published each year in the Spring/Summer edition of the newsletter. The Honor Roll will recognize and honor individuals and corporations for their support of our organization in a cumulative fashion for the previous year.

Development - The CRN fundraising calendar outlines all of the currently identified events for 2008. Additionally, updates from fundraisers held earlier this year are profiled, as well as some new CRN fundraising initiatives. Please take a close look and consider sponsoring an event on our behalf!

Research - The annual Call for Research Proposals occurred in March and submissions are pending – the CRN Scientific Review Board will meet this summer to review submissions and make funding recommendations for new research studies to support. Progress updates for many of the CRN funded studies are provided as well. Much progress has been made and families are encouraged to read these reports closely.

Family Support - In addition to the featured family stories, you will also find information on the 2009 Family Conference, to be held July 16-18, 2009 at the Marriott Evergreen Conference Resort located in Stone Mountain Park, just outside Atlanta, Georgia. The Planning Committee has been working diligently in its pursuit of continuous improvement and is committed to establishing a new standard during the 2009 conference. We will distribute detailed information beginning this Fall.

Education and Awareness - Updates on advocacy activities CRN has performed year to date are included, as well as reports from other cystinosis organizations from around the world. This activity is so critically important to our community and we commit significant resources to ensure cystinosis remains on the forefront of these professional medical organizations.

In closing, CRN published its annual report for 2007 earlier this year – the report can be accessed via the website at <http://cystinosis.org/07annual.pdf>. Major accomplishments in the areas of Research, Family Support, and Education and Awareness are presented, as well as a summarization of CRN's financial profile from the past year. I encourage all to visit the site – we are grateful to everyone whose contributions made all of this possible.

We remain committed to our vision of finding a cure for cystinosis. We ask that you consider becoming involved; from holding a fundraiser, to joining a committee, to attending the Family Conference, you can make a difference.

Please contact me or members of the CRN Board of Directors with any comments or suggestions you may have.

Warmest Regards,

Christy Greeley

President and Executive Director

2009 CRN Family Conference Set For Atlanta



The Cystinosis Research Network is pleased to announce that its 2009 Family Conference will be held July 16-18, 2009 at the Marriott Evergreen Conference Resort located in Stone Mountain Park, just outside Atlanta, Georgia. This will be CRN's fourth family conference, with previous events held in 2003 in Orlando, Florida, 2005 in Salt Lake City, Utah, and 2007 in San Antonio, Texas.

As described on the Marriott's website, "In Frommer's recent travel guide on Atlanta, Stone Mountain Park is described as "one of the most beautiful parks in the country". Visit Evergreen Conference Resort and discover all that this magnificent park has to offer. Stone Mountain, the enormous granite mass featuring the world's largest high relief sculpture is the centerpiece of this 3200 acre paradise. In addition to its stunning natural

beauty, the park offers a wide variety of leisure and recreational activities. No matter your preference, Stone Mountain Park and Evergreen Conference Resort have it all."

More information on the resort and the park can be found on their websites, www.evergreenresort.com and www.stonemountainpark.com.

Please start planning now to join us in 2009 -- Stone Mountain and the Marriott promise to provide a beautiful setting for our families to meet and reconnect. Look for more information regarding conference content and registration on the CRN website at www.cystinosis.org in the coming months.

CRN Financial Update



Alexis LeBeau (7)
-Illinois

"I am very excited to report that 2007 ended on a high note from a financial perspective."

Brittney LeBeau,
CRN Treasurer

By Brittney LeBeau,
Treasurer

Incredibly, almost a year and a half has passed since my transition into the Treasurer position. I am happy to say that job has become much smoother and is nearly running itself...well, almost.

I am very excited to report that 2007 ended on a high note from a financial perspective. CRN collectively raised \$475,000 in income (minus fundraiser expenses). That is the highest fundraising total in CRN history! To view more of the financial details, please go to www.cystinosis.org/07annual.pdf.

To illustrate CRN's financial standing so far in 2008, I have provided the Profit & Loss statement thru February 29, 2008.

Thank you all who have held fundraisers, made personal donations and lobbied for grant money for CRN. I look forward to seeing how 2008 rolls out.

	Jan - Feb 08
Ordinary Income/Expense	
Income	
Direct Public Support	
Individ, Business Contributions	15,963.70
Direct Public Support - Other	3,947.00
Total Direct Public Support	19,910.70
Indirect Public Support	
United Way, CFC Contributions	180.81
Indirect Public Support - Other	3,001.90
Total Indirect Public Support	3,182.71
Investments	
Interest-Savings, Short-term CD	5,381.52
Total Investments	5,381.52
Total Income	28,474.93
Expense	
Business Expenses	5,454.65
Operations	
Postage, Mailing Service	211.75
Printing and Copying	6,000.00
Supplies	26.50
Total Operations	6,238.25
Other Types of Expenses	
Other Costs	567.59
Total Other Types of Expenses	567.59
Travel and Meetings	
Travel	263.80
Total Travel and Meetings	263.80
Total Expense	12,524.29
Net Ordinary Income	15,950.64
Net Income	15,950.64

Sue, Alan, Melissa (29),
Jeremy(21), and
Serena(25 with cystinosis) Scott
—Australia



Frankie, Christopher
(20), and Laura (12
with cystinosis)
McGinnis
—South Carolina

CRN Education and Awareness Update

By Paula Shal, Vice President, Education and Awareness

At the end of 2007 and beginning of 2008, the weather outside was frightful in many parts of the country! Wisconsin was no exception. If I wasn't driving on treacherous roads to and from work every day, I was staying home working on "indoor projects."

As a result, you may have noticed CRN has a new name and design for our newsletter! The CRN Board

reviewed several ideas and ultimately voted for *The Cystinosis Advocate* with a new look that emphasizes the colors in our logo.

The CRN website has undergone a few changes as well. Frequently used links, like the new CRN store and eye-drops resources, have been added to the home page. Mack Maxwell compiled a contact list of doctors who see patients with cystinosis. It is permanently housed in the Resource section.

In addition to using the website and newsletter to

reach out to our community, CRN is busy attending conferences to network, educate others about cystinosis, and receive training to become better advocates. Elva Smith recently attended the National Organization for Rare Disorders (NORD) 25th anniversary gala and International Conference on Rare Diseases and Orphan Drugs (ICORD).

I am looking forward to representing CRN at the Genetic Alliance Conference in July.

More information about this valuable conference can be found on page 19.

"The CRN Board reviewed several ideas and ultimately voted for The Cystinosis Advocate with a new look that emphasizes the colors in our logo."

-Paula Shal

CRN VP Education & Awareness

Cystinosis Research Foundation Update



The Cystinosis Research Foundation held its first international Cystinosis Research Symposium April 3 and 4, 2008 in Irvine, California. Christy Greeley, CRN President and Executive Director, and Elva Smith, CRN Vice President of Research, attended as representatives of the Cystinosis Research Network. The event featured two days of presentations by a variety of new and established clinicians and scientists who are conducting research sponsored fully or in part by CRF.

As noted by Nancy Stack, the goal of the symposium was to ensure that cystinosis research continues to be collaborative and dynamic. CRF

reports that it intends for the symposium to take place every other year.

CRF also recently issued its Spring Call for Research Proposals:

*The Cystinosis Research Foundation is pleased to announce its Spring 2008 call for research proposals and fellowships. We are currently accepting applications for research proposals and for the Post-Doctoral Cystinosis Research Fellowship Program. The CRF currently has over **\$1,200,000** available for cystinosis research and fellowship positions.*

The Cystinosis Research Foundation utilizes a Scientific Review Board comprised of leading experts in the field of cystinosis. The SRB provides independent, objective reviews and recommendations for each research proposal submitted.

The SRB follows grant review guidelines established by the Cystinosis Research Foundation and then advises the CRF on the scientific merits of each proposal.

The Cystinosis Research Foundation gives priority to research that will lead to better treatments and a cure for cystinosis. Both clinical and bench research proposals are accepted. We are especially interested in supporting new investigators and encourage them to apply either as research fellows or investigators.

To learn more about our foundation, visit our web site at www.natalieswish.org or email us at heystack4@aol.com or contact Zoe Solsby at (949) 223-7610.

CRN Research Update



Garrett Thomas (with cystinosis), Mason Stilke (with cystinosis), Rachel Smith, and Mitchell Smith (with cystinosis)

By Elva Smith, Vice President, Research

I am pleased to report that research for improved treatments and ultimately a cure for cystinosis continues its march forward! CRN currently has committed approximately \$750,000 in grant funding, and we're looking to expand this funding as a result of our 2008 Call for Proposals which went out on March 25, 2008.

The eight studies which CRN is currently funding are:

"Early Intervention Trial for Visual Processing Deficit in Cystinosis"

Doris Trauner, M.D., University of California, San Diego, La Jolla, CA

Grant Timeline: August 2003 – ongoing, Award amount: \$112,724

"Development and *in vivo* testing of novel therapies for cystinosis"

Corinne Antignac, M.D., Ph.D., Vasiliki Kalatzis, Ph.D., Hôpital Necker-Enfants Malades, Paris, France,

Grant Timeline: October 2005 – September 2007, Award Amount: \$50,000 Study complete

"Design and Synthesis of Novel Prodrugs for the Treatment of Cystinosis"

Donald Cairns, Ph.D., School of Pharmacy, The Robert Gordon University, Aberdeen, Scotland

Grant Timeline: October 2005 – September 2008, Award Amount: \$97,928

"Tissue Repository for Cystinosis"

Jess G. Thoene, M.D., University of Michigan Medical Center, Ann Arbor, MI

Grant Timeline: Fall 2006 – ongoing, Award Amount: \$26, 206

"Study of ATP metabolism in human cystinotic proximal tubular cells and in humans with cystinosis *in vivo*"

Elena Levtchenko, M.D., Ph.D., University Medical Center Nijmegen, The Netherlands

Grant Timeline: January 2007 – December 2007, Award Amount: \$68,090

"Development of a cysteamine *in situ* gelling system for the topical treatment of corneal crystals in cystinosis"

Dr. C. Tuleu, University of London, School of Pharmacy, Dr. Olufemi Rabi, Guy's & St. Thomas' NHS Foundation Hospital, Mr. Ken K. Nischal, Mr. Rajnish Sekhri, and Dr. William Van't Hoff, Great Ormond Street Hospital and Institute of Child Health

Grant Timeline: Fall 2006-Fall 2009, Award Amount: £ 103,000 (approximately \$203,500)

"Gene transfer studies for cystinosis"

Vasiliki Kalatzis, Ph.D., Eric J. Kremer, Ph.D., Institut Genetique Moleculaire de Montpellier, France

Grant Timeline: Fall 2007 – Fall 2009, Award Amount € 75,900 (approx. \$101,000)

"Evaluation of Novel Prodrugs for the Treatment of Nephropathic Cystinosis"

Professor Donald Cairns, Dr Rachel M Knott, Dr Graeme Kay

Grant Timeline: Fall 2007 - Fall 2008, Award Amount: £ 35,000 (approx. \$70,000)

More detailed information about these grants can be found on the CRN web site www.cystinosis.org

In addition to these grants, CRN has established and funded a three year Cystinosis Fellowship at the National Institutes of Health under the direction of Dr. William Gahl. This will be the first community funded fellowship which will focus on clinical as well as research training in cystinosis, filling a crucial need in our community.

Cystinosis Research Update (cont.)

The following two publications which were partially funded by CRN have recently been made available and can be viewed in full on the CRN website www.cystinosis.org.

A potential new prodrug for the treatment of cystinosis:

Design, synthesis and in-vitro evaluation

Bridgeen McCaughan, Graeme Kay, Rachel M. Knott and Donald Cairns*

School of Pharmacy, The Robert Gordon University, Scotland, Aberdeen, Aberdeenshire AB10 1FR, UK

Received 7 December 2007; revised 11 January 2008; accepted 12 January 2008

Abstract—Nephropathic cystinosis is a rare autosomal recessive disease characterised by raised lysosomal levels of cystine in the cells of most organs. The disorder is treated by regular administration of the aminothioliol, cysteamine, an odiferous and unpleasant tasting compound that along with its metabolites is excreted in breath and sweat, leading to poor patient compliance. In an attempt to improve patient compliance a series of novel prodrugs has been designed and evaluated as a potential new treatment for nephropathic cystinosis. The first of the prodrugs tested, 3a, was found to decrease the levels of intracellular cystine in cystinotic fibroblasts.

This is the first report of a potential new therapeutic treatment for nephropathic cystinosis since the advent of cysteamine bitartrate.

2008 Published by Elsevier Ltd.

Cystine accumulation in the CNS results in severe age-related memory deficits

Tangui Maurice *a,b,c*, Claire Hippert *d,e*, Nicolas Sertratrice *d,e*, Grégoire Dubois *d,e*,

Chantal Jacquet *d,e*, Corinne Antignac *f,g*, Eric J. Kremer *d,e*, Vasiliki Kalatzis *d,e,**

a Inserm U710, 34095 Montpellier, France

b EPHE, 75017 Paris, France

c Université de Montpellier II, 34095 Montpellier, France

d Institut de Génétique Moléculaire de Montpellier, CNRS, 34293 Montpellier, France

e Universités Montpellier 1 & II, 34293 Montpellier, France

f Inserm U574, 75015 Paris, France

g Université Paris Descartes, Faculté de Médecine René Descartes, UMRS574, 75015 Paris, France

Received 11 April 2007; received in revised form 21 August 2007; accepted 18 September 2007

Abstract—Cystinosis is a lysosomal storage disorder characterised by progressive cystine accumulation. The causative gene, *CTNS*, encodes cystinosis, the lysosomal cystine transporter. Neurological deterioration is one of the last symptoms to appear and the least well characterised. Visuospatial memory deficits have been documented in patients. To determine whether the cystinosis mouse model presents similar anomalies, we studied the learning and memory abilities of young and middle-aged *Ctns*^{-/-} mice. We did not detect deficits in young *Ctns*^{-/-} mice. In contrast, spatial reference and working memory deficits were detected in middle-aged *Ctns*^{-/-} mice. Elevated cystine levels were detected in the hippocampus, cerebellum, forebrain and brainstem of all *Ctns*^{-/-} mice, which increased with age and were consistent with the appearance of impairments. Our results strongly suggest that the cystinosis-associated CNS anomalies are due to progressive cystine accumulation. Furthermore, the *Ctns*^{-/-} mice serve as a model to investigate the evolution of these anomalies and test the efficiency of existing and novel treatments to cross the blood-brain barrier and reduce lysosomal cystine levels.

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CRN also received the 24-month, final progress report on a grant initially funded in September 2005. This report may also be seen in full on the CRN website www.cystinosis.org

Development, and *in vivo* testing, of novel therapies for cystinosis

This 2 year research project was a joint proposal between:

Pr. Corinne Antignac (Inserm U574, Hôpital Necker-Enfants Malades, Paris, France) - **Project 1**

Dr. Vasiliki Kalatzis (CNRS UMR 5535, Institut de Génétique Moléculaire, Montpellier, France) - **Project 2**



Abigail Monaghan (2),
diagnosed Oct., 2007
—Ontario, Canada

Call for Research Proposals

The Cystinosis Research Network is pleased to announce its 2008 Call for Research Proposals. Proposals may be submitted to CRN for review and consideration. CRN utilizes a Scientific Review Board comprised of leading experts on the disease of cystinosis which reviews grant proposals and submits funding recommendations to the CRN Executive Committee. More specifically, the SRB provides independent, objective review and recommendations regarding each research proposal utilizing grant review guidelines established

Priority is given to interventional research, both clinical and basic, that will lead to improved treatments for cystinosis. New investigators are particularly encouraged to apply. The Chairperson of the Scientific Review Board summarizes its recommendations and presents them to the CRN Executive Committee, which then votes on each proposed project.

Proposals must be submitted by June 1, 2008. Qualified proposals will be reviewed during the annual Scientific Review Board meeting in early August. The CRN Grant Proposal Guidelines may be accessed on the CRN website at www.cystinosis.org.



Mitomed Offers Cystinosis Gene Testing

The Cystinosis Research Network (CRN) is providing the following information as a service to the community. It is important to note CRN does not have an association in any way with Mitomed Diagnostic Laboratory. Individuals wishing to obtain testing services from Mitomed Diagnostic Laboratory should first consult with their doctor as to the necessity and relevance to their particular situation. Individuals bear the responsibility of determining if testing is covered by their individual insurance. CRN does not provide any financial assistance in this regard.

Service Description: Mitomed Diagnostic Laboratory at University of California, Irvine is now offering genetic testing for CTNS, the gene that causes cystinosis. The lab examines the gene for mutations, which are deleterious changes that affect how the gene works. Genetic testing has a detection rate of 85-90 percent accuracy for people with clinical symptoms. Genetic testing may be helpful in establishing carrier status in unaffected individuals. If possible, it is best to first test the person who has cystinosis.

Once the mutations running in a family are known, it makes carrier testing for other family members easier and more accurate. If the familial mutations are not known, then a negative test result can significantly decrease a person's carrier risk in relation to the general population. Genetic testing may also be helpful in prenatal diagnosis and pre-implantation genetics (PGD). Prenatal diagnosis can be arranged by our laboratory through GeneDx.

Mitomed has three board certified geneticists, a board certified PhD geneticist and two genetic counselors on staff. They are more than happy to give information to the medical community and patients over the telephone with respect to testing strategy and interpretation of test results. Families in need of in person genetic counseling services are welcome to schedule visits through the UCI clinical service. They are also able to recommend colleagues at other locations.

Please contact Mitomed Diagnostic Laboratory at 949-824-1886 if you have questions regarding genetic testing for cystinosis and their services. More information, including requisitions and sample requirements, can be found on their website: <http://mitomed.bio.uci.edu>.

Participate in Cystinosis Research



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To patients and parents of children with cystinosis:

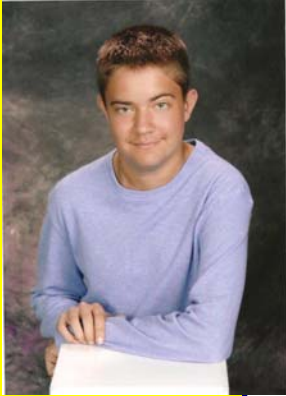
A tissue repository for renal tissue from patients with cystinosis is being established, with funding from CRN, at the University of Michigan. The existence of this repository will enable ongoing study of cystinotic tissues, which is currently hindered by lack of available tissue.

If you want to participate, please request that appropriate tissue samples be forwarded by the pathologist who analyzed you or your child's renal tissue, whether obtained at renal biopsy during management of the disease, or at transplantation, and who has custody of the sample, to the repository at the University of Michigan. The samples will be maintained confidentially. The identity of each patient will be unknown to University of Michigan Repository staff, but will be marked with an identifier so that samples can be retrieved if future clinical needs dictate. Acceptable samples include unstained slides, or portions of paraffin blocks, depending on the amount of tissue available. All donations must comply with applicable hospital and State laws and requirements.

If you are interested, please ask your nephrologist or pathologist (if known) to contact Dr Jess Thoene at the University of Michigan, Division of Pediatric Genetics, 734-272-5573 who will cover details of sample coding and clinical summary to preserve confidentiality and ensure appropriate shipping. Please do not contact Dr. Thoene directly, as this will violate confidentiality.

Funds are available to cover the costs of shipping of the specimen.

The Cystinosis Research Network is an all-volunteer, non-profit 501(C)3 organization, Federal Tax ID # 04-3323789.



Shea Hammond
(senior picture)
—Massachusetts

Participate in Cystinosis Research

We would like to encourage everyone to keep in mind the importance of research in our conquest of cystinosis. When you and your children participate in a cystinosis research study, you are helping to improve the quality of life for all of those affected by cystinosis. Without your help, many research studies may be impeded, and none of us want that. Please consider what your contribution can be to make the CRN goal of the discovery of improved treatments and ultimately a cure for cystinosis a reality.

The Pediatric Neurology Research Group at the University of California, San Diego (UCSD) is conducting the following research study on cystinosis: Academic Achievement in Cystinosis and Cystic Fibrosis

The UCSD Pediatric Neurology Research Group is conducting a study to examine academic achievement, which includes skills such as reading, writing, and arithmetic, in individuals with cystinosis or cystic fibrosis. Participants will include individuals with cystinosis or cystic fibrosis ages 5 years through 18 years. This study will involve approximately three 2- to 2½-hour cognitive-behavioral testing sessions. The testing will help us learn more about academic achievement in cystinosis and cystic fibrosis. Participants and/or teachers will also be asked to fill out questionnaires regarding school behaviors. Participation in this research study is completely voluntary.

If you would like to participate in the Academic Achievement study or if you would like more information, please contact Kirsten Kung at UCSD: kpoehlmann@ucsd.edu or 858-822-6800.

UCSD's Cognitive and Neural Development Study

*How does your brain choose important sights and sounds from the environment?
What goes wrong with cystinosis?*

We are looking for healthy individuals, as well as those with cystinosis, 6 to 45 years of age, from all language backgrounds.

What is the study about?

This study is to help us understand how the auditory and visual systems function in people with cystinosis and what the origin of late-onset brain damage is.

How do we conduct the study?

There are two 2-hour sessions, during which we record the participant's brain waves while they "play a computer game." In one session, the participant looks at a computer screen and presses a button once a certain image is presented in a designated location. During another session, the participants will attend to sounds presented through several speakers and will press a button every time they hear a specific sound. Recording the activity of the brain is non-invasive, completely painless, and safe. (In rare cases, there may be some mild skin irritation). This method is widely used in child and adult research. The participants wear a special hat with the sensors of electrical brain activity plugged into it. Our participants sit in a comfortable chair in a soundproof booth while playing the game. The identity of you or your child, as well as of all data records, is entirely confidential.

What are the benefits of participation?

For participating, children and adults will receive \$20 per session. Children will also receive a small toy as incentive.

How do I schedule a session?

To schedule a session or for more information, please contact:

Mikeala at (858) 822-1961, mkinnear@ucsd.edu

Applied Physics and Mathematics bldg. Prime

9500 Gilman Dr.

LaJolla, CA 92093-0113

<http://www.crl.ucsd.edu/pcnd>

Participate in Cystinosis Research

Blood Sample Preparation for White Blood Cell Cystine Levels

Dr. Jerry A. Schneider and Dr. Meredith C. Fidler are conducting a research study to learn more about how to prepare the blood samples used for the determination of white blood cell cystine levels. For this study, they are looking for cystinosis patients (12 years and older) who would travel to San Diego for this study. Flight cost will be paid for. Preference will be given to patients 18 years and older, who can fly to San Diego and back home on the same day. Patients do not have to currently be taking Cystagon. However, patients have to be able to provide recent laboratory test showing that their red blood cell count is not below normal.

White blood cell cystine levels need to be measured occasionally to be certain patients are taking the correct dose of Cystagon. When we started to study cysteamine many years ago we tried to find a way to have whole blood shipped to our laboratory so that we could both prepare the white blood cells and measure the cystine. We couldn't make this work. We only got reliable results if the white blood cells were made promptly after the blood was drawn.

Although preparing the white blood cells is not very difficult, it is very time consuming and labs often find the preparation challenging, especially if a lab only does this every few months. We have some ideas of how we might stabilize the white blood cells so that the blood could be shipped and our laboratory could prepare the white blood cells. To test these ideas we need some cystinosis patients who are willing to donate blood. Patients volunteering for this study will be asked to come to the UCSD (University of California, San Diego) Medical Center in San Diego where a blood sample will be taken. Depending on the age and blood count of the patient, up to 10 tablespoons of blood will be taken (150 mL). A "unit" of blood that is taken at a blood bank is three times as much (450 mL). Of course smaller patients will have less blood taken. Patients may be asked to not take Cystagon for 24 hours before the blood draw. Participating in research is entirely voluntary. If you are interested in participating and would like more information, please contact Meredith Fidler, PhD, at (619) 543 2049 or mfidler@ucsd.edu. 200 West Arbor Drive, MC 8450 San Diego, CA 92103-8450 Phone (619) 543 2049

CRN Patient Volunteer Listing

The Cystinosis Research Network is compiling a listing of individuals who are interested in participating in cystinosis research studies. If you are interested in having your name added to this list, you will be asked for some basic contact information and you will give CRN permission to release this information directly to researchers who are looking for individuals with cystinosis and/or their family members to participate in research studies. The researchers may then contact you directly in the future with various research opportunities. Submitting this

information in no way indicates your commitment to participate in any clinical trial. Rather, it simply gives your permission for researchers to contact you directly.

If you would like to have your name added to the listing, please complete the following:

Patient's Name:

Date of Birth:

Parents' Names:

Address:

Phone:

Email:

This information can be mailed or emailed to:

Elva Smith

Vice President, Research

417 Clairemont Avenue

Unit 209

Decatur, GA 30030

elvasmith@mindspring.com

By forwarding this information, you give CRN permission to release it to researchers and their staff. You may have your name removed from this list at any time by contacting Elva Smith at the above address.

The Southwest Foundation for Biomedical Research Takes "Big Picture" Approach to Cystinosis Research

A Call for a Cure

"I just crumbled," says Kim Shepperd as she recalls hearing the news that her second child would be born with cystinosis. All she and her husband, John, could think of that day was how the onslaught of the disease had ravaged their son, John Ben, and how living with cystinosis had changed their lives exponentially.

Neither of them was thinking of the old adage quoted in the *Sound of Music*: "Where God closes a door, somewhere he opens a window." But now they say that's what happened just a few weeks later.

It was late on a Friday afternoon, and Kim's father, Richard Azar, was about to leave his office when a courtesy call came from Southwest Foundation for Biomedical Research (SFBR), an organization he and his wife, Dianne, had long supported. Development associate Amy Abdalla wanted to thank the Azars for a recent donation, and a short conversation ensued.

Suddenly Azar thought to ask, "Do you all do any research on cystinosis?" He and his wife admired this San Antonio-based independent biomedical research organization and the strides it was making in the fight against maladies such as heart disease, diabetes, obesity, infectious diseases, mental illness, and other diseases that impact premature infants to the elderly. Now he was hoping the organi-

zation might help defeat an orphan disease that had twice hit his family.

After that phone call, Abdalla followed up with SFBR's Genetics Department, where scientists are known for their expertise in hunting down genetic contributions to common complex diseases such as heart disease and diabetes.

The response she got? The department didn't have any research projects on cystinosis, but Dr. John Blangero – who is well known internationally for his innovative methods in statistical genetics – had a "never-been-done-before" idea that he wanted to try. He thought SFBR's extraordinary genetic resources and expertise were well suited for an entirely new approach to studying a monogenic disease.

"With our traditional focus on complex diseases, which involve multiple genes and environmental factors, we tend to look at problems globally," says Dr. Blangero. "So we decided to turn this problem on its head and ask, 'What could we do with our genetic approaches and resources that would inform us about the [biological] causal pathway of cystinosis?' In other words, we wanted to learn more about the biology of this disease and how the *CTNS* gene actually functions – since there is very little science on this – as well as what other genes 'upstream' impact *CTNS*, what other genes *CTNS* impacts 'downstream', and what other genes might perform parallel, or similar, functions to *CTNS* so that we

could look for new drug targets and other areas of disease intervention," he explains.

"We believe the findings from this type of study could relate not only to cystinosis, but also to other diseases, including those caused by a similar toxic build-up of amino acids, such as Alzheimer's and Parkinson's. And if this research method proves effective in the study of cystinosis, it could become the new paradigm for the study of other monogenic, or single-gene, disorders," Dr. Blangero says.

A series of meetings was set up with the Azars, Shepperds, and SFBR representatives. Dr. Blangero learned more about the family's plight, and they listened to his novel research proposal, which also involved a stellar molecular genetics group that had recently moved to SFBR from Australia, led by Drs. Eric Moses and John Blangero.

The Azars were so impressed by the potential of this big-picture approach that they provided the initial funding for the creation of a new cystinosis research program at SFBR, and they have since helped the program maintain its momentum with supplemental philanthropic grants. Their financial support has been augmented by more than \$850,000 in generous contributions from others, including two separate grants totaling more than \$330,000 from the Cystinosis Research Foundation – Natalie's Wish.



Photos of the Shepperd children serve as inspiration for SFBR scientists as they conduct their research.



The SFBR cystinosis research team includes (counterclockwise beginning at bottom right) Drs. John Blangero, Eric Moses, Matthew Johnson, Katy Freed and Joanne Curran.

Continued from page 12

Other substantial donations have come from the Elizabeth Huth Coates Charitable Foundation, the Southwest Foundation Forum, the Lanward Foundation, and numerous relatives and friends who have made individual gifts.

The payoff has been impressive, with SFBR investigators embarking on several "firsts" in cystinosis research and garnering some exciting results.

A new approach to single-gene disorders

One of their first steps was to learn more about the normal function and variation of the *CTNS* gene in a random, unaffected population so they could learn more about what the gene does in a healthy individual. That would lend insight into what goes wrong when the gene is disrupted.

This research approach had never been applied to the study of a monogenic disorder, nor had it been used in such a large-scale analysis. In what Dr. Blangero describes as "the single largest transcriptional profiling project ever undertaken," Dr. Moses' team performed genome-wide transcriptional scans on 1240 members of SFBR's San Antonio Family Heart Study.

What does that mean? In short, genes work by producing transcripts, or messenger RNAs, which then are turned into proteins that have particular biological effects. So the SFBR team performed genetic tests to measure the mRNA output level of all the genes in the genome of each individual studied.

At the same time, they sequenced every piece of DNA in the *CTNS* gene of 200 individuals and found the normal, common variations in the gene among individuals. "In other words, we found variations in the DNA sequence of that gene that have big effects on its output," says Dr. Blangero.

Large-scale study produces large-scale results

Then they analyzed this massive amount of data and found more than 2,000 genes that correlate with *CTNS*, making them the subject of follow-up studies to see if they might be good targets for disease intervention.

Some of those appear to be genes that are "downstream," or genes that are affected by the output of *CTNS*. Researchers say these could potentially be good targets for treating disease symptoms or complications.

Other genes of interest are believed to be "upstream" from *CTNS*, meaning they seem to impact its function. Dr. Blangero explains, "For the 30 to 40 percent of cystinosis cases in which the *CTNS* gene is disrupted but still functioning, these genes 'upstream' in the biological pathway might be used to help regulate *CTNS* and improve its function."

Dr. Moses says the research team is following up on one such gene that strongly correlates with *CTNS* and that other research has shown to play a role in "cleaning out" damaging chemicals from cells. That could make it a target in fighting multiple diseases.

Other identified genes, the researchers say, appear to perform similar, or parallel functions, to *CTNS*. They're following up on one in particular after interest was sparked from an unlikely source: yeast.

Researchers at another institution found that when the yeast equivalent of *CTNS* was "knocked out," another gene essentially "stepped up" and performed a similar function. With this exciting information, Drs. Blangero and Moses used the human genome sequence and the genome transcriptional profiles they had developed and identified one matching human gene, or homologue, to this newly identified yeast gene.

"That's one of the things that we're working on right now," says Dr. Blangero. "We want to see what this gene does and if we can manipulate it. There is a big evolutionary distance between yeast and humans, so all the gene's functions might not be preserved crossing from one to the other, but we want to see if we can manipulate this gene in such a way that it will take the place of *CTNS* in people with cystinosis."

Pursuing hot leads

For follow-up studies on this promising homologue, as well as some of the other genes they've identified to be of greatest interest, SFBR researchers are now embarking on some new collaborations and methods of research.

One developing collaboration is with French nephrologist Dr. Corinne Antignac, who first discovered the *CTNS* gene. In her research at *Hôpital Necker-Enfants Malades*, a children's hospital in Paris, Dr. Antignac has developed a mouse model in which the *CTNS* gene has been knocked out.

Investigations with this model can help study the relationship of *CTNS* and the yeast homologue and other genes of interest to researchers, reveal more about these genes' functions, and help show if and how those functions change when *CTNS* is disrupted. One of the grants provided by the Cystinosis Research Foundation is helping support this vital effort.

SFBR researchers also got some help from the Cystinosis Research Network. When the group hosted its international conference in San Antonio in July 2007, SFBR researchers used the opportunity to recruit 150 members of affected families to donate blood samples for further study.

Continued on page 14



Dr. Matthew Johnson makes use of a new genetic sequencer that is beneficial in SFBR's research on cystinosis and other diseases.

"SFBR researchers also got some help from the Cystinosis Research Network.

When the group hosted its international conference in San Antonio in July 2007, SFBR researchers used the opportunity to recruit 150 members of affected families to donate blood samples for further study."

Researchers have since used these 150 samples – obtained from children with cystinosis as well as from their parents and siblings – to develop "immortalized cell lines" for a variety of complementary research projects, supported in part by a second grant from the Cystinosis Research Foundation.

"Right now, we're sequencing these individuals' *CTNS* genes and performing genome-wide expression profiling with each individual for comparison studies with our unaffected population," says Dr. Moses. "With this information, we'll be able to learn more about all the different variations of *CTNS* and how they correlate with varying degrees of severity of cystinosis. We will also learn more about the effect of these mutations on the genes downstream in the biological pathway.

"And really exciting is that we'll be able to conduct a variety of tests in these cell lines to see how we might manipulate genes upstream, downstream and parallel to *CTNS*, and how those genetic manipulations impact cystine metabolism, or the cells' ability to clear out excess cystine. For example, we can flood a cell with high levels of cystine, then do something to over-express one of these *CTNS* correlates and see what effect it has. Is the gene we have our eye on truly a good drug target or not?"

A serendipitous discovery

In a separate but related effort, SFBR scientists used a large set of genetic profiles developed through their cystinosis research in a new method they developed for genetic discovery. Then they used that method to find a gene – *VNN1* – that regulates levels of HDL, the "good" cholesterol. Now, serendipitously, the *VNN1* discovery could lead to an exciting breakthrough in the treatment of cystinosis and many other diseases.

VNN1 is known to produce cysteamine, which helps transport excess cystine out of cells and prevents it from accumulating to toxic levels. Cysteamine also removes other dangerous things from cells, including excess glutamine, which can lead to Huntington's and Alzheimer's diseases, and general oxidative stress, which plays a key role in heart disease.

The new findings about *VNN1* by SFBR scientists are expected to give pharmaceutical companies good reason to pursue drugs that could increase the gene's activity, stimulating it to produce more cysteamine naturally. That could result in new preventions and treatments for a variety of disorders, and it could reduce the need for cystinosis patients to take cysteamine orally.

Moving forward

These exciting developments gave Dr. Katy Freed and her mentor, Dr. Eric Moses, good news to report at the Cystinosis

Research Foundation's First International Cystinosis Research Symposium, held April 3-4, 2008. Dr. Freed, a post-doctoral scientist on the SFBR cystinosis research team, is funded by one of the foundation's grants.

They also propel the group forward to what they hope will be even more exciting discoveries – and some critical federal funding for cystinosis research. Until now, SFBR's approach would have been too novel to be considered for a grant from the National Institutes of Health, especially since it focused on an orphan disease.

However, with the tremendous strides made over the past few years and the great potential to positively impact the lives of people with cystinosis as well as other human health disorders, SFBR scientists believe they now have enough compelling data to merit consideration for NIH funding. They plan to submit a federal grant application for the cystinosis project by the end of 2008.

That payoff could be huge. "We have so many leads to explore, so many opportunities to make a significant contribution to the fight against cystinosis," says Dr. Moses. "And we truly believe our novel research methodology will be the new paradigm for the study of single-gene disorders. It could revolutionize this field of research."

Article and photos provided courtesy of Southwest Foundation for Biomedical Research in San Antonio, Texas.

Meet Christian Morales



Christian Morales

By José and Velyna Morales

May 6, 1994 was truly a glorious day. That was the day Christian was born. We were blessed with a son to nurture and raise in God's house. Like all parents, we felt our child would have limitless potential to achieve so many things....we had ***the greatest expectations for our son.***

Everything proceeded as one would expect. Parents focused on every

need of their first born and made every effort to ensure the health and well-being of their child. Things went well in the beginning, but soon symptoms began to emerge that would take us on a journey of discovery. Christian was finally diagnosed with cystinosis in 1999 by Doctor Jerry Schneider. Once Christian was confirmed to have cystinosis, we dedicated ourselves to learning everything there was about the disease and to exact as much influence over our destiny as humanly possible. On May 20, 2004, Christian received a kidney transplant....Velyna was his donor..."a mother bear taking care of her cub".

As we write this article, we cannot help but reflect on all the victories we have celebrated since that day of diagnosis. Christian is a dedicated and diligent student. His commitment in pursuing his dreams is amazing....whether it is playing the piano, achieving academic excellence in the classroom, or living out his dreams of athletic feats.....he always gives his all. Describing the true essence of Christian is a challenge:

- Christian is a sensitive and compassionate individual. Christian is always ready to reach out to those less fortunate. He recently participated in a 30 hour famine to raise money and food for the hungry.
- Christian is courageous and sets a standard for us in terms of leadership. Christian does not complain about the heavy regimen of medicine and doctor appointments. He is always ready to participate in studies which will benefit others....which invariably means even more medicine and more doctor appointments.

- Christian is a teenager facing all of life's changes. Christian endeavors to do the right thing and find his place in his circle of friends.....learning to live with his body.
- Christian is a believer and endeavors to live his life in a Christ like fashion. Christian is very active in his church and youth group

It would be less than genuine if we did not confess there are challenging times; times of discord and passionate debates. The way we see it, this is simply a part of life and growing up.

Christian is our son, our hero. We give thanks to God for sharing him with us as he teaches us on a daily basis to celebrate all of God's gifts and to give thanks for the life we all share.

Today, more so than ever, we have ***the greatest expectations for our son.***



José and Christian Morales



Christian, José, Velyna, and Alexandra Morales

Miss United States 2007 Ashley Kazian Spreads the Word About Cystinosis



MISS UNITED STATES 2007
Ashley Kazian

www.Cystinosis.org

Ashley Kazian's cards include CRN's website address



MISS UNITED STATES 2007
Ashley Kazian

www.Cystinosis.org



Dr. Gahl got crowned at the NIH

Wow! I can't believe my year as Miss United States is already half over. I have enjoyed every minute, every appearance, every obstacle, and every person I have met along the way. Being the Cystinosis Spokesperson has allowed me to travel around the country talking about this rare disease and raising awareness. While traveling on "official duty" I am required to wear my banner in the airport. As a result, I have met some of the most amazing people in airports and have had the opportunity to share the stories of the Cystinosis Patients. Every autograph card that I sign has the Cystinosis web site on it, as do my business cards. I also carry brochures with me that explain the disease and how I got involved with the Cystinosis Research Network.

In December, I had the opportunity to travel with Frankie and Laura McGinnis to The National Institutes of Health in Bethesda, Maryland. We stayed at the Children's Inn where we interacted with children from around the world with rare diseases. I am always amazed by their positive attitudes and loving spirits. While at NIH, I had the privilege of meeting Dr. Gahl and his dedicated staff. I was able to go with Laura to her doctors appointments. As I sat there watching her get poked, prodded, and drilled with questions, I realized that my love and admiration for this young lady grows each time I am around her. Laura and I were interviewed on a local television show called "Your Carolina" in January, where we discussed Cystinosis and how the community can get involved.

Because of the last newsletter, I have received many emails from those involved with Cystinosis and I am excited to be a part of two charitable events coming up. On May the 4th, I will fly to Bloomfield Hills, Michigan for the 2nd annual 5k Cystinosis Run. The Wyman family has worked around the clock to make this fundraiser a success, and I cannot wait to attend. Dana Dury is hosting "A Festival of Hope" on September 18th in Downers Grove, IL. I am honored to fly out for this event, and cannot wait to see how much money is raised for research.

I get hits daily on my personal MySpace account which is covered with Cystinosis information. Many people are curious, as most have never heard of the disease. As frustrating as it can be to not be able to have an instant solution, spreading the word and raising awareness is what it is all about. I would love to hear any stories, or fundraising ideas, so please do not hesitate to contact me.

Thank you to the entire Cystinosis community for allowing me to be a part of this special family.

Much Love,

Ashley Kazian

Miss United States 2007

www.MySpace.com/ashleykazian

Amkzn@aol.com

864-346-6539



Ashley and Laura enjoy some down time at the NIH



Ashley Kazian and Laura McGinnis appeared on "Your Carolina" with Jack Roper and Kimberly Kelley in January

Mikaela Gard's Wish Comes True at American Girl Place



Mikaela poses with her American Girl Doll at the American Girl Place Cafe

Mikaela Gard took a trip to American Girl Place-Chicago in June, 2007 through Kid Wish Network. Mikaela's trip included a stay at the Westin Hotel on Michigan avenue as well as meals at the Rainforest Café, Buca di Beppo, and the American Girl Place cafe.

At lunch in the American Girl Place Café, Mikaela was given the royal treatment. Her doll had a booster seat and place settings, and Mikaela had a personal shopper who brought her a gift.

She also received a shopping spree at American Girl Place. Her personal shopper gave her a tour of the store and then left her to shop its three levels. The personal shopper told Mikaela to fill her shopping basket and come get her when it was full.

"Mikaela shopped til' she dropped—literally," said Mikaela's mom, Dena. When she got back to the hotel, she went straight to bed and didn't even look at her new loot until she woke up."

Dena said Mikaela made some lasting memories that will last a long time.

"She was in heaven."

Mikaela was diagnosed with cystinosis at age 10 months.

She is now 14. She has gone through everything from rickets to GI problems. Although her creatinine is currently 1.1 (It was .7 one year ago), Dena said Mikaela is doing great.

"Today she is so stable it is scary," said Dena.



Dena and Mikaela Gard at Buca di Beppo in Chicago

"Mikaela shopped til' she dropped—literally. When she got back to the hotel, she went straight to bed and didn't even look at her new loot until she woke up."

—Dena Gard



MEXICAN ASSOCIATION OF CYSTINOSIS ANNOUNCEMENT

5th Medical Cystinosis Symposium
will be held in Mexico City
January 2009

Complete information and agenda coming soon at:

www.cystinosismexico.org

info@cystinosismexico.org

Upcoming Family and Medical Events

May 20, 2008

NORD 25th Anniversary Gala / ICORD Conference
Washington D.C.

June 27-28, 2008

International Cystinosis Conference
Dublin, Ireland

July 4-6, 2008

Midwest Cystinosis Gathering
Casey, IL

July 11-13, 2008

Cocoa Beach Cystinosis Gathering
Cocoa Beach, FL

July 11-13, 2008

Genetic Alliance Conference
Bethesda, MD

July 11-16, 2008

NKF Transplant Games
Pittsburgh, PA

September, 2008

NORD Annual Conference
Washington D.C.

November 4-9, 2008

American Society of Nephrology
Philadelphia, PA

January, 2009

5th Medical Cystinosis Symposium
Mexico City, Mexico

July 16-18, 2009

2009 CRN Family Conference
Atlanta, GA

August 29-September 2, 2010

Fifteenth Congress of the IPNA
New York, NY

July, 2011

2011 CRN Family Conference
San Francisco, CA

Family Gatherings Set for July

Cystinosis Family Gatherings will take place in Florida and Illinois during the month of July. A family gathering is an opportunity network with other cystinosis families in a fun and informal setting. There are no doctors or presentations.

These gatherings are open to any family affected by cystinosis from all over the world. You do not have to live in the region they are being held.

The Midwest Family Gathering will be held on July 4-6 at the Comfort Inn, 933 N. State Hwy 49, Casey, IL. Reservations can be made by calling 217-932-2212.

A family gathering will take place at the Hilton in Cocoa Beach, Florida July 11-13. Room rates are \$129 per night, and reservations can be made at www.hiltoncocoabeach.com.

Refrigerators are included in some rooms or are available for a small fee.

Rooms can be upgraded to an ocean view for an additional charge. Cocoa Beach is just 45 minutes from the Orlando airport. The Kennedy Space Center is just a few minutes from the hotel. E-mail Sandy Glaize at Sandy-Glaize@aol.com with questions.

More information on both gatherings will be at www.cystinosis.org as it becomes available.

Fifteenth Congress of the International Pediatric Nephrology Association (IPNA) Scheduled for 2010

By José Morales

The Fifteenth Congress of the International Pediatric Nephrology Association (IPNA) will convene in New York City from August 29 – September 2, 2010. Dr. Fredrick Kaskel has been the designated the host member for this exciting event. The Congress will join 1,200 to 1,500 pediatric nephrologists and investigators from all over the world.

Consistent with IPNA's long-standing history of commitment to clinical excellence, education, research and training, the 15th Congress will build upon what has been achieved in the past and bring together the most advanced investigators in the fields of basic and clinical nephrology. The Congress will facilitate the interchange of new information to expand and en-

hance the understanding of normal and pathophysiologic basic or renal disorders critical to the field of pediatric nephrology. A primary goal of the Congress is to provide the framework for continuing education of the pediatric nephrology community at all levels with a special emphasis on trainees and new members of the IPNA. The Congress' agenda will be built around specific themes to generate collaboration and enhance the community's understanding of genetic renal disorders: *Cystinosis*, *oxalosis*, *Alport's syndrome*, *polycystic kidney disease*, etc.

The International Pediatric Nephrology Association is providing major organizations the opportunity to sponsor the Congress. Access and recognition will be commensurate with the level of sponsorship. As the designated lead non-

profit organization and the important nature of the Congress, CRN sponsorship will be at the premium level. CRN has formed a Planning Committee consisting of Chairs José Morales and Marybeth Kruppenacker to lead our efforts. More specifically, CRN views the Congress as a great way of further educating IPNA members about cystinosis.

CRN will be collaborating closely with the professional meeting planner (Convention Headquarters Inc.) in providing input and assistance as necessary regarding activities specifically related to cystinosis. A website is under development and will be online soon to provide greater details: www.IPNA2010.org.

CRN to be Represented at Genetic Alliance Conference



Jim and Paula Shal will represent CRN at the Genetic Alliance Conference in Bethesda, MD, July 11-13. The conference will kick off with a Genetics Day on the Hill July 10.

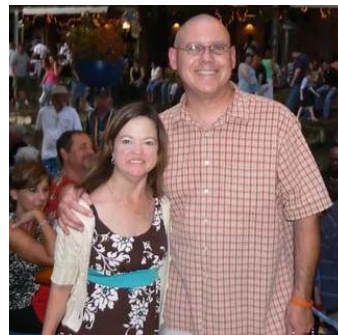
Genetic Alliance is a coalition of more than 600 advocacy organizations serving 25 million people affected by 1000 conditions. The organization works to transform leadership in the genetics community to build capacity in advocacy organizations and to educate policymakers by leveraging the

voices of individuals and families. Genetic Alliance increases the capacity of genetic advocacy organizations to achieve their missions and leverages the voices of millions of individuals and families living with genetic conditions.

Genetic Alliance is dedicated to improving the quality of life for everyone living with genetic conditions. The Genetic Alliance Conference is designed to train advocacy leaders on various topics that will benefit advocacy groups such as CRN.

There will be daylong symposia on topics such Leadership, Organization, Research, and Policy. There will also be smaller workshops on topics such as genetic testing, ethical genetic technol-

ogy, maintaining medical data fact sheets, newborn screening, patient empowerment, nutrition, insurance coverage, and more. In addition, there will be many opportunities to network with other advocacy leaders. Look for a synopsis of this conference on the CRN website and in a future edition of *The Cystinosis Advocate*.



Paula and Jim Shal

“Genetic Alliance is dedicated to improving the quality of life for everyone living with genetic conditions.”

National Kidney Foundation Hosts U.S. Transplant Games

The National Kidney Foundation invites all transplant recipients, living donors, and donor families to join in the NKF Transplant Games, July 11-16th in Pittsburgh, PA.

The Transplant Games are a biannual event in which recipients of organ transplants compete. The event brings together recipients, living donors and donor families to show the world that transplantation works!

Competition is open to anyone who has received a life-

saving organ transplant—heart, liver, lung, kidney, pancreas. Bone marrow recipients are also eligible to participate. As much as the Games are an athletic competition that calls attention to the success of organ and tissue transplantation, it is also a celebration of life among recipients, their families and friends.

The Games demonstrate to the public the collective and individual successes of the life-restoring therapy of organ transplantation. The Games use the mass media

to promote the success of organ donation and transplantation and to call attention to the need for organ donation through events and support activities before, during and after the event.

Some individuals with cystinosis routinely participate in the Games, including Barry Beard and CRN Secretary, Karen Gledhill.

If you are interested in attending the Games, please contact your local National Kidney Foundation to find out about the team in your area.

Development Update



Tyler (9) and Megan (11-with cystinosis) Morrill
-Michigan

By Jill Morrill, Vice President, Development

I hope this newsletter finds you all enjoying Spring, staying healthy, and thinking about fundraising for CRN. First, I must thank all of you who have raised money or contributed to an event for our organization. We directly depend on your generous donations and time to keep our researchers and doctors working hard and our cystinosis families as healthy as possible. Huge thanks to all of you.

We have had seven successful fund raisers so far this year and if you look on the calendar, we have more to look forward to. However, we still need your help to fulfill our commitments. As the development coordinator, I am here to cheer you on and encourage you all to try raising money.

Not all fundraisers have to be time consuming. Below are some great ideas:

- Personal donation
- Letter campaign (just ask)
- Garage sales and lemonade stands
- Girls/guys/couples night out (book club, card group, poker night, just a get together)
- Spring fling
- Home shows (jewelry, candles, cookware, home decor)
- Shop at www.cafepress.com/cystinosis
- Company matching gift programs
- United Way, GoodSearch, Ebay
- Block party or backyard barbecue

- Refer to our fundraising calendar for more ideas such as Biking, Golfing, Carnivals, Hoedowns, Birthday Parties, etc...

I also found that if you just Google an idea on the computer, loads of organized step by step "how to run your fund raiser" are right at your fingertips. Google "a-thon fundraisers" and a whole new world opens up. It is actually fun and inspirational to see how much can be achieved if we all pitch in and go for it. Any amount of money raised is wonderful. It all adds up.

Remember, you can always contact me at jill.m.morrill@gmail.com I am here for all of you in any way possible.

Make an Online Donation Through PayPal!



Did you know that you can make a secure online credit card donation to the Cystinosis Research Network through PayPal?

- You can dedicate a donation to someone specific. PayPal passes along your dedication to CRN.
- You will receive a record of your contribution for tax purposes.
- Go to www.cystinosis.org and click on the "Donate" link to begin!

2008 Fundraising Calendar

Date	Event	Location	In Honor Of
Ongoing	CRN Note Cards	www.cystinosis.org/cystinosisorder-form021005.pdf	N/A
Ongoing	GoodSearch.com	www.goodsearch.com	N/A
Ongoing	CRN Logo Merchandise	www.cafepress.com	N/A
Ongoing	Fundraising Letter Campaign	N/A	Mason & Livia Stilke
January 18	"Shoot for the Cure" Pancake Supper	Whitesboro, TX	Mason Reed
March	Jack Greeley's Birthday Fundraising Letter	N/A	Jack Greeley
April	Stilke's Partylite Fundraiser	N/A	Mason and Livia Stilke
April	Ukrop's Monument 10K Run	N/A	Alex Weaver
April 12	Lowdown at the Hoedown	San Antonio, TX	John Ben and Ava Shepperd
May 3	5K Fun Run	Bloomfield Hills, MI	Kacy Wyman
May 16	3rd Annual Golf Tournament	Berthoud, CO	Neveah Stanford
June	A. Joseph DeNucci Golf Tournament	Newton, MA	Shea Hammond
June 7	Cycle for Cystinosis	Dekalb, IL www.afestivalofhope.org/otherevents.html	Alexis and Evan LeBeau
June 28	2nd Annual Gerry Veles Golf Outing	Mundelien, IL	Jack Greeley
August 7	16th Annual Herb Didier Memorial Golf Outing	Lincolnshire, IL www.golfinvite.com/lincolnshirerotarygolf	Jack Greeley
September 6	C.H. Robinson 2nd Annual Cystinosis Golf Tournament	Lehi, UT	Tahnje Woodward
September 18	A Festival of Hope	Downer's Grove, IL www.afestivalofhope.org	Alexis and Evan LeBeau
October	Silpada Jewelry Fundraiser	www.mysilpada.com/jill.morrill	Megan Morrill
October	Wyman Holiday Letter Campaign	N/A	Kacy Wyman

Jack Greeley's 8th Birthday Letter Campaign



Jack and Dave Greeley

The Greeley Family of Chicago, Illinois has reached out to family, friends and colleagues for the sixth year and asked for donations to be made to CRN in honor of their son Jack's 8th birthday on March 17. The Greeleys have raised over \$125,000 with their letter campaigns, with last year's letter raising approximately \$23,000. This year's total is still pending as donations continue to be received.

The theme of this year's letter focused on the incredible generosity and support that the family and CRN has received,

told through the examples of two teenagers who chose to contribute to CRN in honor of Jack over the past year:

"While we started writing this letter a few years ago to simply gain financial support for the fledgling CRN to help our son and others, we have learned that we have received a truly special opportunity from Jack's plight. It is said that to whom much is given, much is expected. For us, Cystinosis has also become an instrument to touch the lives of others; to truly give others pause to reflect and appreciate

what they have and what they can do in life. It has become a gift of perspective and appreciation for us to share and for others to embrace..... Our journey with Cystinosis has shown us how much good still exists in today's complex world. We have chosen to embrace it and we invite you to embrace life too, no matter what it has to offer."

Read this year's inspiring letter by visiting www.cystinosis.org/news031008.pdf and those from past years by visiting www.cystinosis.org/news031707.html.

Maddie Parmacek Chooses CRN for her Mitzvah Project

My name is Maddie Parmacek. On October 27, 2007, I was called to the Torah for my Bat Mitzvah. Becoming a Bat Mitzvah took a year of hard work and dedication. The next day, my parents threw me a party where all my friends and family helped me celebrate my achievement. When a Jewish child has a Bat Mitzvah, they are asked to choose a Mitzvah project. The term Mitzvah means to do something good. For the Mitzvah Project, we are asked to find something that interests us and figure out a way to help some person or group that is in need. For my Mitzvah project, I chose to take a portion of my money that I received as gifts for my Bar Mitzvah, and make a donation to the Cystinosis Research Network. I chose the Cystinosis Research Network after my sister and family told me about Jack Greeley's courage and continued perseverance with this condition. Knowing the special relationship my sister and my family has with the Greeley family, my decision in choosing to help the Cystinosis Research Network in any way I could was an easy one. I hope my donation can help to make a difference in the lives of Jack and other children who have this condition.



Maddie Parmacek and Jack Greeley

Thank you for fundraising!

- Cuts for Kacy raised over \$2,000 in honor of Kacy Wyman!
- Jill Morrill's Silpada Jewelry fundraiser raised \$235 in honor of Megan Morrill!
- Ukrop's Monument 10K Run raised \$1,245 in honor of Alex Weaver!
- Lucas and Shane Wysocki's birthday party raised \$2,250 in honor of Jacob Ellerbock!
- The Stilke's Partylite Fundraiser and letter campaign raised \$1,850 in honor of Mason and Liva Stilke!
- The 5K Fun Run raised \$30,000 in honor of Kacy Wyman!

Look for more information about these and other fundraisers on the CRN website and in the next issue of *The Cystinosis Advocate*.

“Shoot for the Cure” Honors Mason Reed



On January 18, 2008, the Whitesboro boy’s basketball program hosted a pancake dinner to raise money for the Cystinosis Research Network. The dinner was part of the “Shoot for the Cure” night in honor of Mason Reed, nephew of Head Coach Shad Reed.

Mason Reed with his uncle, Shad Reed, and Aunt, Keva Reed

The Whitesboro boys also honored Mason as an honorary captain during the game. He was introduced into the starting lineup and given an autographed basketball. After that, he headed to the bench to help his uncle do some coaching. The night was a huge success with over \$1,000 being raised for this worthy cause.

“It was great to see such a huge amount of support for my family. We have great kids and a great community

that are willing to help out in any way possible. I’m proud to be associated with this community and I’m very proud of the kids in our program,” said Shad Reed .

Whitesboro is located about one hour north of Dallas, Texas. Mason resides in Bushland, Texas and is the son of Ryan and Kylene Reed. He attends kindergarten at Bushland Elementary where he is one of the top students in his class. He is also active in baseball, soccer, and basketball.

“It was great to see such a huge amount of support for my family . We have great kids and a great community that are willing to help out in any way possible.”
—Shad Reed

3rd Lowdown at the Hoedown Doesn’t Disappoint

By John Shepperd

The third Lowdown at the Hoedown was held in April this year at the historic Anhalt dancehall about 25 miles North of San Antonio. We didn’t hold the fundraiser in 2007 because it would have fallen so close to the Family Conference, so our volunteers were eager to get back into the swing of things. The weather was perfect, and we had a good turnout. We were excited to see some new faces this year, due mainly to the article on the Hoedown that appeared in the San Antonio newspaper a couple days before the event.

The kids activities are the highlight of the afternoon....moon bounce, Wiggle-Waggle train, face painting, temporary tattoos, and the petting zoo. The “train” is actually a large riding lawn mower towing several brightly colored, cut-out 55 gallon barrels. Our volunteers really stepped up this year, cooking all the food

themselves, rather than paying a caterer. One group stayed up almost all night tending the barbecue smoker, and the effort really paid off. We cut our food expenses by about 75%, and it was absolutely terrific.

Another highlight was the artwork donated by Kevin McCalla. His charcoal drawings were outstanding, and we really appreciate his support!

I don’t know the final tally, but I think we raised somewhere in the neighborhood of \$8000 for CRN. We were lucky enough to find sponsors for all the major expenses, so then it’s up to us and the volunteers to make it all happen. I encourage ALL of you to hold a fundraiser of some type. It is a lot of fun, and as you know, CRN counts on support from its membership. Don’t be afraid to hit up your friends and neighbors for help. People want to get involved, but sometimes they just don’t know how to start.



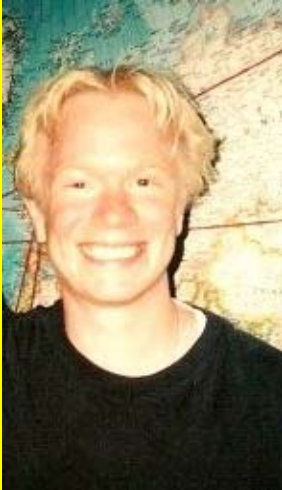
Mason Reed and the Whitesboro Boy's Basketball Team



John Ben Shepperd at the Lowdown at the Hoedown



Ava and Kim Shepperd at the Lowdown at the Hoedown



Steve Schleuder—
2007 Scholarship
Recipient

2008 CRN Scholarship for Individuals with Cystinosis

The Cystinosis Research Network has established a scholarship fund to provide supplemental financial assistance to a student diagnosed with Cystinosis who is enrolling in a regionally accredited collegiate or vocational program, or who is currently attending a post – secondary school. The scholarship award, \$1000, is awarded contingent upon the winner’s acceptance to an accredited college, university, or vocational program, or documentation of continued enrollment, and will be payable to the educational institution to be applied toward tuition.

An application form is available on the CRN website at www.cystinosis.org. For more information, please contact: CRN at 1-866-276-3669 or crn@cystinosis.org.

APPLICATION PROCEDURE:

Each applicant must submit:

1. Documentation/verification of Cystinosis (e.g. Letter from physician)
2. An official copy of high school transcript
3. Two letters of recommendation from current teachers/faculty members and/or counselors regarding applicant’s scholastic aptitude and personal qualifications.
4. An essay of 500 words discussing the applicant’s personal and educational goals

JUDGING CRITERIA: The essay will be judged on the basis of rationale, grammar, and comprehension. Transcripts and letters of recommendation will be considered in the final decision with Grade Point Average (GPA), courses taken, and class standing used as part of the evaluation. The Cystinosis Research Network Board will establish an independent judging panel to evaluate and rate the applicants. The decisions of the judges are final.

Finalists may be interviewed before selections are made.

DEADLINE FOR APPLICATION: Application and all accompanying documents must be received at the Cystinosis Research Network office postmarked **by August 15, 2008**. FAXES OR E-MAILS WILL NOT BE ACCEPTED.

PREPARING APPLICATION PACKAGE: Each application packet must include a complete application (original or photocopied—go to www.cystinosis.org to download), required documentation materials and essay, on 8 1/2” X11” white paper. Send all materials in a single, flat package. All application documents become the property of the evaluation committee.

MAIL APPLICATION PACKETS TO:

Sandy Glaize
4133 Conway Place Circle
Orlando, Florida 32812

2008 CRN Sierra Woodward Sibling Scholarship

The Cystinosis Research Network has established a scholarship fund to provide supplemental financial assistance to a student who has a sibling diagnosed with Cystinosis who is enrolling in a regionally accredited collegiate or vocational program, or who is currently attending a post – secondary school. The scholarship award, \$1000, is awarded contingent upon the winner's acceptance to an accredited college, university, or vocational program, or documentation of continued enrollment, and will be payable to the educational institution to be applied toward tuition.

An application form is available on the CRN website at www.cystinosis.org. For more information or to have an application mailed to you, please contact:

Marybeth Krummenacker at MKRUMM@aol.com or 516-931-6785 or

Pam Woodward at rptswood@csolutions.net or 801-404-6119

APPLICATION PROCEDURE:

Each applicant must submit:

- 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 300 to 500 words discussing the applicant's personal and educational goals and also the pro and/or cons of how Cystinosis has affected your life

JUDGING CRITERIA: The essay will be judged on the basis of rationale, grammar, and comprehension. Transcripts and letters of recommendation will be considered in the final decision with Grade Point Average (GPA), courses taken, and class standing used as part of the evaluation. The Cystinosis Research Network Board will establish an independent judging panel to evaluate and rate the applicants. The decisions of the judges are final.

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MAIL APPLICATION PACKETS TO:

Marybeth Krummenacker

54 Smith St

Hicksville, NY 11801



Tony Prankienas (2),
diagnosed May, 2007
-Michigan

Join the Cystinosis Research Network

Get connected! Stay informed! Together we can find a cure!

Join The Cystinosis Research Network (CRN) and become part of a global network of caring families, concerned individuals and healthcare professionals working together in the fight against cystinosis. The Cystinosis Research Network's vision is the discovery of improved treatments and ultimately a cure for cystinosis. The CRN is an all-volunteer; non-profit organization dedicated to sponsor and advocate research, provide family support, and educate the public and medical communities about cystinosis. The CRN funds research and programs primarily through donations from the public, grassroots fundraising events and grants.

CRN provides outreach and access to resources. We take great pride in carrying out our motto:

"Searching For A Cure"...whether you are ...

- A **Parent** who needs critical resource information, support services or help in sharing the challenges of cystinosis to those who serve your child.
- An **Adult** with cystinosis interested in information regarding medical and social issues that are specifically geared for adults.
- A **Relative** or a **Friend** who wants to increase their understanding of cystinosis and find out how you can help out or become involved.
- A **Physician, Social Worker, Educator** or other **Professional** who makes a difference in the life of a family affected by cystinosis, and want to have access to critical information to better serve your patient, student or client.

Joining the Cystinosis Research Network enables you to:

- Receive all the latest cystinosis information through our countless resources, including the CRN Newsletter, our very informative web page www.cystinosis.org, the popular online Cystinosis Support Group, and our toll free number (1-866-276-3669).
- Attend the CRN Family Conference with other cystinosis families to exchange knowledge and create friendships. Also, find out the latest discoveries about cystinosis from the medical professionals.
- Let your voice be heard by legislators and policymakers who need to know why cystinosis (and other rare diseases) are important issues to you.
- Have access to the Cystinosis Research Network's representatives in the areas that are most relevant at any given time to you or your loved one affected by Cystinosis.

Join the Cystinosis Research Network today!

Thank you for your consideration in becoming a member of the Cystinosis Research Network.

Pam Woodward

VP Family Support, Cystinosis Research Network

Cystinosis Research Network Membership Form:

Immediate Family:.....\$20.00	Extended Family / Friend:....\$25.00
Professional:.....\$35.00	International: (Including Canada) Base rate (see above categories) plus \$10.00 for postage. Payable in US dollars

Please complete the form & mail with check payable to CRN to:
 Cystinosis Research Network
 302 Whytegate Ct.
 Lake Forest, IL 60045

Name _____
 Street _____
 City & State _____ Zip Code _____ Country _____
 Phone _____ Fax _____ Email _____

Name of Child / Adult / Acquaintance / Patient affected with cystinosis:

Join A CRN Support Group

Looking for a way to communicate with others in the Cystinosis

Community on a day-to-day basis?

The Cystinosis Research Network offers two email support groups for communicating with others in the cystinosis community:

The CRN Support Group is a group for parents, affected adults, caregivers, family, and friends. We also welcome researchers and medical professionals who are interested in cystinosis. This is the place to discuss the various aspects of cystinosis, and how it affects our lives, how we cope, vent our frustrations, share our fears, our hopes, and our dreams.

The CRN Teen Support Group is for teens with cystinosis and teenage siblings of children and adults with cystinosis. Connect with other teenagers who are dealing with similar issues. The posts include questions, concerns, ideas and supportive sharing.

To join a support group, visit www.cystinosis.org/support.html.



Amelia Douglas (with cystinosis) married Douglas Miller in August, 2007

Please Support CRN's Mission with Your Donation

YES, I want to help children and adults with cystinosis.

Enclosed is my tax deductible contribution of: \$ _____ made payable to the Cystinosis Research Network (CRN) and mail to: 302 Whytegate Ave., Lake Forest, IL 60045

Name _____

Street _____

City & State _____ Zip Code _____

Phone _____ Fax _____ Email _____

In Honor Of _____

In Memory Of _____

You may send notification of my gift to:

Please check all that apply:

____ Friend

____ Individual with Cystinosis

____ Parent of Child with Cystinosis

____ Professional

____ Family

____ I am interested in volunteering for CRN. Please contact me.

Search the Internet Using GoodSearch and Raise Money for CRN

Every time you use GoodSearch.com to search the Internet, a donation is made to the Cystinosis Research Network! GoodSearch.com is powered by Yahoo!, so you get the same great results you get from most search engines. What is unique is that GoodSearch.com has developed a way to direct money to your selected charity with every click!

To get started, go to www.goodsearch.com. Be sure to select "Cystinosis Research Network" where it says "Who do you GoodSearch for?" Then search like you normally would! It costs you nothing.

CRN hopes you will give it a try and support Cystinosis Research. The more people who use this site for CRN, the more money is earned. So please tell your friends and family!

Donate to CRN by Selling on eBay

CRN is registered with MissionFish, the exclusive charity provider for eBay Giving Works. eBay sellers can now list items through eBay Giving Works and designate a percentage of the sales to go to CRN. The seller picks the percentage, and all money donated is tax deductible. eBay will even refund a percentage of listing and final value fees that is equal to the percentage sellers donate! Items listed with eBay Giving Works are given a special icon, so they stand out. Some sellers report 20-40% higher sale prices for the exact same item using eBay Giving Works. Give it a try, and be sure to tell established eBay sellers about this great opportunity to give to CRN!





United Way Contribution Guidelines

Identify the Cystinosis Research Network, Inc. as the agency you want to receive your contribution through the United Way Donor Choice Program.

Agency Name	The Cystinosis Research Network, Inc.
Non-Profit Tax ID #	04-3323789
Address	302 Whytegate Ct., Lake Forest, IL 60045
Telephone	1-866-276-3669 (toll free), 1-847-735-0471
Fax	847-235-2773
E-mail Address	CRN@cystinosis.org
Web Page	www.cystinosis.org

The local United Way organization will contact The Cystinosis Research Network via phone, fax, or e-mail to request we prepare and submit documentation verifying our status as a non-profit organization.

The Cystinosis Research Network prepares all necessary documentation and submits it to the respective local United Way organization.

The local United Way organization processes the documentation and sends a check for the aggregate sum designated for the Cystinosis Research Network.

The Cystinosis Research Network sends thank you/acknowledgement letters to recognize contributing individuals.

Note Card Fundraiser

CRN is selling lovely note cards to support cystinosis research. Give a gift that says you care and also supports a worthy cause.

Design options are script initials, floral, "thank you," and more. The inside is blank. The CRN logo and website address is on the back. A set of 8 cards and envelopes is \$10.00 (US), which includes shipping and handling.

Go to www.cystinosis.org/fundraisers.html to download an order form.

Buy Cystinosis Research Network Gear at the CRN Store

CRN has teamed up with cafepress.com to offer CRN merchandise. Items include t-shirts, sweatshirts, hoodies, tanks, camis, hats, bags, housewares, buttons, printed material, and more! Clothing is available for men, women, children, and even pets! The best part is that up to 30% of your purchase goes back to CRN!

Some items are available in bulk. Clothing and accessories are great to wear to cystinosis fundraisers.

Access the CRN Store at:

<http://www.cafepress.com/cystinosis>

Shop til' your heart's content, and be sure to tell your friends and family!





Laura McGinnis (12)
-South Carolina

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*Amounts denoted are monies received to the best of our knowledge from January 1, 2007 through December 31, 2007 and may not include cash donations or gifts in kind, etc.

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Erik (2)
-Australia
(and Stubby!)

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Kenadee Julian (5)
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Continued from page 1

"They would be ready to throw up as soon as they saw those meds coming," she said.

Because they just could not keep those meds down, it created problems with growth, abnormal labs, and many hospitalizations.

"Their size has always been something that makes them feel out of place in school. Missing so much school caused them to struggle academically as well."

Rachel says staying on top of the medicine has been a particular struggle with having twins.

"They were on the same meds but different doses. I had to be careful not to give them the wrong doses."

Things also got pretty hectic around the time Anji started dialysis, and Nelly was not on it yet. Rachel ran back and forth from the clinic to school, ran errands, and had to try to pick them up around the same time at opposite ends of the city.

"Someone usually had to wait," Rachel said.

Media Coverage

The local media got involved in Anji and Nelly's story in mid-October, 2007—prior to their transplants. Rachel was first aware that the girls' story was groundbreaking when public affairs said they wanted to interview her and get a story.



Anji and Nelly with their mom, Rachel

"Before that, I just thought it would be a normal thing for twins that needed transplants," said Rachel.

Just about every news station covered the story as well as The Chicago Tribune, Chicago Sun Times, and The NBC Today Show.

Rachel appreciates the opportunities that were given to her family to share their story. She hopes she helped the cystinosis community in some way.

Positive Attitudes

The girls are doing great since their transplants and have not had any abnormal labs.

After having the transplants in Chicago, Rachel and the girls have moved to Greenwood, Indiana. Rachel is engaged and expecting another child in October. The girls will start fifth grade in the Fall. They have always been creative and love art. They also like playing with their friends and their puppy. They are big fans of the shows *iCarly*, *Hannah Montana*, and the *Wizards of Waverly Place*.

Rachel believes a positive attitude and laughter has helped the family cope with the challenges of cystinosis.

"As with any child with cystinosis, they are very brave. Their sense of humor is wonderful, and I believe that because we learned to laugh in the face of this illness, we were able to enjoy every minute of every day, no matter how bad things got."



Cystinosis Research Network

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Editor: Paula Shal

CRN Vision and Mission

Vision. *The Cystinosis Research Network's vision is the discovery of improved treatments and ultimately a cure for cystinosis.*

Mission. *The Cystinosis Research Network is a volunteer, non-profit organization dedicated to supporting and advocating research, providing family assistance and educating the public and medical communities about cystinosis.*