



## Why I Love the International Cystinosis Congress

By Michaela Drury

**W**hy do I love the International Cystinosis Congress so much? This is a question I often get asked by people who have never attended a congress when they are trying to decide whether it is worth saving up and taking the time to travel and attend an International Cystinosis Congress. I have been to three; Paris, Manchester and Valencia and I can honestly say the answer is most definitely yes!

The first congress I went to was in Paris and I so wish I had gone sooner. The first time I heard about the congress was a couple years before when I saw people talking about the 2010 Italy congress however my family does not like to travel and I was too scared to go on my own as I had never been on a holiday abroad before. I remember seeing all the pictures on Facebook afterwards and feeling so sad that I was not there. So in 2012 when I saw the Paris conference and my new partner agreed to come with me, I went for it and have been going ever since.



MATTHEW MENCE

L to R: Michaela Drury, Holly Paine, Ami Froelich and Freek Wonnink enjoy the night life at the conference.

It is very difficult to put into words for someone who has never been exactly why I get so much out of them. I will try to tell you about the latest International Cystinosis Congress that I attended in Valencia Spain this past July as a way of explaining why this conference is so important to me.

I saw the 9th International Cystinosis Congress Beyond Borders, Valencia, Spain on the Cystinosis Foundation Facebook page and at once started to make plans. I looked at the price of flights to Valencia and accommodation rates. I decided

[SEE CONGRESS page 7]

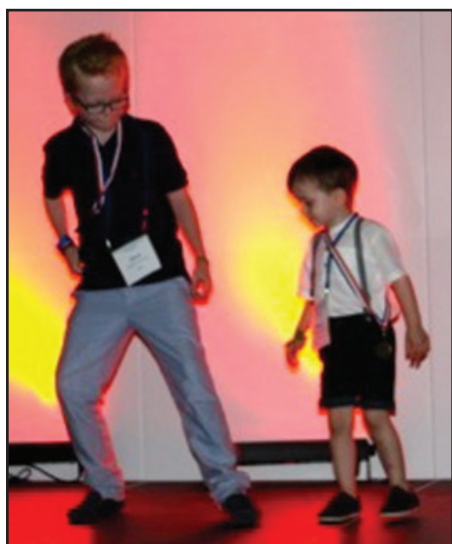
## 9th International Cystinosis Congress Salutes Our True Heroes

By Merle Mund

**T**he Cystinosis Foundation hosted a wonderful conference in Valencia, Spain this past summer from Thursday, the 30th of June to Sunday, July 3rd. Valencia is a beautiful city situated right on the Mediterranean Sea with many ancient and modern attractions for families to visit after the conference. The soft breezes from the sea contributed to the lovely climate in Valencia.

Lourdes Sanz, President of Grupo Cistinosis Espana, co-hosted this gathering of 228 people. She shared that about 60 people affected by Cystinosis live in Spain. We are grateful for the guidance and support of our scientific chairs, Gema

[SEE NINTH page 10]



Everyone enjoyed dancing on Saturday night.

### INSIDE

	Page
Amazon SMILE	21
Cystinosis Bone Meeting	4
Cystinosis Foundation Membership	23
Donations	20-21
Faces in the Crowd	12 -14
Fertility in Male Cystinosis Patients	4
From the President	2
Our Heroes and Trailblazers	5
Hobbs Humanitarian Award	3
Horizon Pharma	6
In-Kind Gifts	23
In Loving Memory	
Annie Fencil	16
David Reuter	17
Brian K. Riddle	18
Krista Mund	19
Photos from Valencia Conference	8-9
Potts Scholarship	22
Thank you	6, 7
What is Cystinosis?	3
Your Gift Saves Lives	20

# From the President's Desk

By Jean Hobbs-Hotz

**D**ear Friends, some of you may recall in 1999 – as we transitioned from the 20th to the 21st century – the editors of TIME Magazine cast a backward glance and named Albert Einstein the Person of the Century. Considered the greatest mind of the 20th century for his many profound contributions to science, his ideas influenced our modern culture in many other ways.

When I think of our mission that we have been pursuing since 1983, to make a difference for the child born today, I think of what Albert Einstein once observed,

“A hundred times every day I remind myself that my inner and outer life depends on the labors of other men, living and dead, and that I must exert myself in order to give in the measure as I have received and am still receiving.”

We cannot accomplish anything of any real value alone and by ourselves. If we can ever accomplish anything, it is the result of collaboration with other like-minded people. It is the great value of coming together that the Cystinosis Foundation and its members have understood and shared since the very beginning.

Holding fast to this concept has led us to help establish 15 cystinosis patient support groups in 15 different countries around the world. Our sense of service and collaborative spirit has banished isolation and created a truly global community of patients and families who are there for each other.

At the 9th International Cystinosis Congress in Valencia this summer we honored our true heroes and trailblazers, those

who have departed and those of us who continue on this journey together. We have been graced over the years with the friendship of outstanding individuals who have given back to their community in order to benefit all. This is service in the fullest meaning of the word.

We honor Freek Wonnink and Joshua Hotz, recipients of the W. L. and Sophia Hobbs Humanitarian Award for their contributions to furthering scientific understanding of cystinosis, so that all may eventually benefit from advancements in research. I think of Dr. Albert Schweitzer, who wrote, “I don’t know what your destiny will be, but one thing I do know: the only ones among you who will be really happy are those who have sought and found how to serve.” This is the call of the Cystinosis Foundation and we are most grateful to Freek, Joshua and the many others who have given of themselves.

Dr. Schweitzer worked in the direct service of humanity by bringing healthcare to Gabon, which was, at that time, a remote part of the world. In addition to building a hospital there, in 1952 he used the money he received as a result of winning the Nobel Peace Prize to build a new building for those suffering from leprosy.

He was more than a physician – he was a great humanitarian who wanted to alleviate suffering – caring for thousands and healing sick people who otherwise would have died. He did so despite the intervention of two world wars, disease and his



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Jean Hobbs-Hotz

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own old age.

With the holidays and a New Year upon

[SEE PRESIDENT page 15]



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

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

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



# Hobbs Humanitarian Award Recognizes Wonnink and Hotz

By Valerie Hotz

The Cystinosis Foundation honors outstanding individuals who give back to our community by presenting the W.L. and Sophia Hobbs Humanitarian Award. Prior to 1983, families and individuals living with cystinosis were alone, extremely isolated, with nowhere to turn. No patient support group existed then to serve the seemingly insurmountable needs, most crucial at the time of diagnosis.

W.L. and Sophia shared a passionate commitment to the well being of others, not only within their own family, but also within their greater community and their daily behavior exemplified this loving kindness. With the help of friends, their daughter, Jean Hobbs Hotz, created the Cystinosis Foundation, a 501 (C) (3) non-profit charitable organization serving all families coping with cystinosis. It is only fitting that the Hobbs Humanitarian Award be presented to members of our international community who exemplify loving

kindness in their daily lives and practice giving back to our community.

This award embodies the words of Albert Einstein, who said, “A hundred times every day I remind myself that my inner and outer life depends on the labors of other men, living and dead, and that I must exert myself in order to give in the measure as I have received and am still receiving.”

The 2016 Hobbs Humanitarian Award recognizes the contributions of Freek Wonnink and Joshua Hotz for their thoughtfulness and compassionate giving of themselves to advance scientific understanding of cystinosis.

Freek worked for his local government in the Netherlands in information technology, eventually successfully establishing his own software consulting firm. Throughout his teens and early twenties, Freek did not walk. After his physician explained that his condition was not preventing him from walking, Freek began



Left to Right: Freek Wonnink and Gianni Fogliada.

the tedious, difficult and extremely challenging process of physical therapy to regain his mobility. Throughout Freek has participated in medical studies, often at great pain and discomfort to himself, to increase medical knowledge about cystinosis, thereby benefiting all cystinosis patients.

Joshua Hotz is a musician and artist who enjoys playing piano and guitar, although the muscle wasting he experiences prevents him from playing guitar. In spite of having suffered vision loss ten years ago, Joshua carves exquisite walking sticks. After his diagnosis in 1982 at the age of 15 months, his grandmother, Jean

[SEE HOBBS page 6]



Merle Mund, member of the Board of Directors of the Cystinosis Foundation presents the 2016 Hobbs Humanitarian Award to Joshua Hotz. First Row: Joshua Hotz Left to Right: Merle Mund, Valerie Hotz and Timothy Hotz.

## What is Cystinosis?

Cystinosis is an ultra rare metabolic disease that results in the abnormal accumulation of the amino acid cystine in essentially all of the cells, tissues and organs of people affected by the disease. As this amino acid accumulates it forms crystals in the cells. These cystine crystals damage critical organs including the kidneys, the brain, the pancreas and the eyes. Without treatment, patients typically succumb to renal failure by approximately ten years of age.

## Fertility Research in Male Cystinosis Patients

By Elena Levchenko, MD, Ph.D.

Hormones are small signaling molecules that regulate metabolism, growth and development, and the function of several other tissues in the body. The glands producing these hormones are called the endocrine organs. In cystinosis, various endocrine organs can get affected. For example, in the second decade of life, the function of the thyroid gland gets impaired in many patients. Later on, also the pancreas may show difficulty in producing insulin and enzymes involved in digestion.

The testicles in males and the ovaries in females, called the gonadal organs or gonads, are also endocrine organs. These glands produce the sexual hormones, which are responsible for the development of some of the sexual characteristics (development of body hair and the growth of the penis in males and breasts in females throughout puberty), and for reproduction (conception and pregnancy). Involvement of the gonadal organs due to cystinosis is primarily a problem in male cystinosis patients.

Until now, no male cystinosis patient has been able to induce pregnancy spontaneously. It is known that in most male cystinosis patients, the testicles show difficulty in producing reproductive hormones (e.g. testosterone). This primary testicular malfunction, called primary hypogonadism, may be due to the progressive accumula-



Elena Levchenko, MD.

tion of cystine crystals, inflammation and the subsequent development of scar tissue within the testicles. It frequently results in the delay of puberty, which is observed in male cystinosis patients.

Next to hormonal dysfunction, recent research has shown that in all male cystinosis patients there is a complete absence of sperm cells (spermatozoa) in the ejacu-

late. This is called azoospermia. This absence of sperm cells is the main cause of infertility in male cystinosis patients. On the other hand, the production of these sperm cells in the testicles themselves (spermatogenesis) has shown to be intact when a piece of testicular tissue was examined. Today, it remains unknown why no spermatozoa are present in the sperm of male cystinosis patients. Further research is needed to unravel this problem.

Fortunately, technology in reproductive medicine has advanced, creating opportunities for couples struggling with infertility. Nowadays sperm cells can be harvested from the testicles or epididymis of a male patient, and preserved at low temperature for a certain time needed. Subsequently, successful conception is feasible, even with one sperm cell only. The technology that is used to induce conception with only one sperm cell is called intra-cytoplasmic sperm injection (ICSI). This technology has become available in several countries around the world and it could satisfy one of the most important needs in life of our male cystinosis patients. Hopefully, in the near future this standard of care could be provided to our cystinosis patients, through an extensive multidisciplinary collaboration between the treating physician, a fertility specialist, urologist and gynaecologist, in recognized and experienced fertility centers around the globe.

## 2016 Cystinosis Bone Meeting Takes Place in Austria

Cystinosis patients have many risk factors for fragile bones, including loss of phosphorus in the urine, chronic kidney disease, thyroid disease, low muscle strength and abnormal vitamin D metabolism.

The 2016 Cystinosis Bone Meeting takes place in Salzburg, Austria at the Schlosswitz in Anif from Thursday, December 8th to Saturday, December 10th. Organized and hosted by Katharina Hohenfellner, MD, Head of Pediatric Nephrology in the Kliniken Südostbayern AG, Traunstein, in association with Cystinose-Selbsthilfe e.V., the express goal is to develop guidelines for diagnosing, treating and monitoring bone disease in cystinosis.

This interdisciplinary meeting will host patient support groups from Europe and the United States to hear specialists from 21 different countries deliver presentations on endocrinology, pharmacology, pediatric nephrology, pathology, radiology, orthopedics, physiotherapy and pediatric neurology. The program is designed for scientists to brainstorm together to create potential solutions for patients' needs to develop stronger bones.

"We are thrilled to have the participation of many key opinion leaders contributing to this important meeting," says Hohenfellner. Participating cystinosis experts include Dieter Haffner, MD, Pro-

[SEE BONE page 11]



Katharina Hohenfellner, MD and Cystinose Selbsthilfe, e.V. host the Bone Meeting this month.



# Heroes and Trailblazers of the Cystinosis Community

By Don Hammond

Cystinosis means a journey on a new frontier. I have personally been a part of the cystinosis community since 1984. I am an observer and admirer of the international Cystinosis Foundation. I have been tremendously inspired by each patient and his or her family and stand in awe of the physicians and researchers who have dedicated themselves to the exploration and increased understanding of this condition and to helping patients.

I have watched those with Cystinosis, their families and the skilled experts, move from being a handful of individuals, who were unconnected, to being a united global community that helps one another and has effective treatment options.

1983 was the early frontier of understanding cystinosis. It was vast and unknown. A grandmother, Jean Hobbs Hotz, along with several of her friends, including Sue Russell, Ellen Armstrong, Wendy Clark, along with parents Merle and Bruce Mund, Karen and Frank Ritchie, Gayle and Ernest Britt, who had children who had been diagnosed and came together when Jean's grandson, Joshua, was diagnosed in 1982. Understandably overwhelmed with lack of understanding and lack of resources, it was the beginning of the cystinosis community. They came

together for mutual support and moved to organize other families facing this frontier together. They discovered even more families and soon learned this condition affected children and families around the world.

These early organizers and participants were reluctant heroes. They became leaders and foundation founders because of their children's needs and the needs of others who felt alone and overwhelmed. They had no great financial resources. What they did have was great compassion.

Little did they know how their actions would build an international community. Today there are far more resources to aid families and individuals who are forging ahead on the frontier of improved health and quality of life for those with cystinosis and those who support them. Every culture has pioneers and heroes. It is important we remember and honor those pioneers on the journey toward understanding and improving life in the cystinosis frontier

Sarah Melang has passed away. Sarah wrote, "Living with a chronic illness has its good days and bad days. But I have learned to value each moment of every day, to enjoy the little things in life, a sunset, a hug, a smile and the sound of a young child laughing."

It is important to remember there was



For over 30 years Don Hammond has participated in Cystinosis Foundation programs, often playing his acoustic guitar for everyone's enjoyment.

no treatment for cystinosis until the late 1970's and even then it was only experimental. Cystagon was not approved until 1994. Helen and Robert Fenstermacher lost three children to Cystinosis, losing two children within two weeks of one another. There had been no treatment for their children. Dr. Schneider reports with great courage Ruth was the first child to take a dose of cysteamine as a treatment for cystinosis. They did not know what the outcome would be. But Ruth and her parents were willing to try. Ruth met her husband, Dean Heinzerling, who also had cystinosis, at our cystinosis conference in La Jolla, California. Dean's sister, Heidi, was a Cystinosis patient as well.

Krista Mund became the second individual to take a dose of cysteamine as a treatment for cystinosis. She spent nine weeks in the hospital and by doing so, she helped scientists adjust the proper dose for patients over time. We are grateful to them for their contributions and to the many others who continue to help us blaze this trail. The participation of so many patients in numerous studies has contributed to our understanding of cystinosis today.

Our history sets the stage for the continuing journey as we embrace our past

[SEE HAMMOND page 15]



Don leading a youth encounter session at the 2010 conference in Lignano Sabbiadoro, Italy.

# Maker of Procysbi Acquired by Horizon Pharma

In a press release issued from Dublin, Ireland on October 25, 2016, Horizon Pharma announced it has completed the acquisition of Raptor Pharmaceuticals, Inc. Timothy P. Walbert, chairman, president and chief executive officer of Horizon Pharma reports, “The acquisition of Raptor directly aligns with our long-term strategy and evolution into a rare disease focused company, where now more than half of our medicines are used to treat patients with rare diseases.” Horizon Pharma currently markets 11 medications, with six of those being rare disease treatments.

With a U.S. office based in Lake Forest, Illinois, Horizon Pharma’s press release describes the firm as a biopharmaceutical company “focused on improving patients’ lives by identifying, developing, acquiring and commercializing differentiated and accessible medicines that address unmet medical needs.”



Claudia Sproedt of Cystinose-Selbsthilfe e.V. and Kristina Broadbelt, Director of Patient Advocacy for Horizon Pharma, at the 9th International Cystinosis Congress in Valencia, Spain.

Many of our families and members know Kristina Broadbelt, Director of Patient Advocacy for Raptor Pharmaceuticals, who remains in her post as a Director of Patient Advocacy for Horizon Pharma.

According to Matt Flesch, Executive Director, Product Communications at Horizon Pharma, the company will continue the patient support services Raptor had in place. “This includes disease education programs, access and reimbursement services where appropriate, as well as active support of patient advocacy organizations like the Cystinosis Foundation,” says Flesch.

The Cystinosis Foundation has a strong record over the past 33 years of building effective collaborative relationships with several industry partners to help serve our global community and looks forward to partnering with Horizon Pharma.

For more information about Horizon, go to [www.HorizonPharma.com](http://www.HorizonPharma.com). Send your questions by email to [Connect@HorizonPharma.com](mailto:Connect@HorizonPharma.com).

## ◆ HOBBS from page 3

Hobbs-Hotz, established the Cystinosis Foundation for all children and all families affected by this condition. Joshua has, to use Einstein’s words, “exerted himself in order to give in the measure as he has received...”, by participating in countless scientific studies to benefit scientific knowledge of cystinosis.

We admire the kindness and compassion shared by Freek and Joshua. Their contributions go above and beyond and it is with deep gratitude that the 2016 W. L. and Sophia Hobbs Humanitarian Award is presented to Freek Wonnink and Joshua Hotz.

## Making A Difference Since 1983 For the Child Born Today

The University of Michigan  
Medical School  
Department of Pediatrics  
Division of Biochemical Genetics and Metabolism

Jess G. Thoene, M.D.  
Professor and Chief of Service

Ronald Holmes, M.D.  
Associate Professor  
Rosemary Lemons  
Research Associate

March 27, 1997

Jean Hobbs-Hotz  
President Cystinosis Foundation  
2516 Stockbridge Drive  
Oakland, CA 94611-2413

Dear Jean:

My thanks to you and all the members of the Cystinosis Foundation for your continued support of our research into cystinosis at the University of Michigan. As you know, you are the only funding source at the moment and therefore your continued support is vital to our research efforts.

Thank you again. I look forward to seeing you all at the annual meeting in July.

Sincerely yours,

A handwritten signature in dark ink, appearing to read 'Jess'.

Jess G. Thoene, M.D.



## ◆ CONGRESS from page 1

as I would be in another country I would like to stay at the conference hotel and use the cystinosis congress discount code that was available. There were other budget hotels within 5 mins walk, but when going abroad I like to stay somewhere nice and nearly everyone attending the conference would be staying at this same hotel.

Having realised the congress was in school time - I work at a primary school - I would have to ask for a few days leave, which I knew was technically not allowed during school term. Since I was determined to go, I printed off the event dates and agenda with a covering letter explaining to the head teacher why I wanted to go and how I felt it would benefit me. I had worked out that with travel time and my low energy levels and fatigue it would be best to arrive the day before so that I could have a good night sleep and get to know the area and where to get water and food before the conference started.

So a few days later when I was in class and got handed a letter by a school receptionist you can imagine my surprise when I opened the letter to find my application for leave had been refused, stating that leave is only permitted for medical and training purposes and that the conference did not count as either. This was very upsetting. To start with I decided 'oh well I can't go' and that was that. However, I then spoke to other cystinosis patients who encouraged me to appeal. I wrote to HR with supporting letters from the Cystinosis Foundation and my doctors. About a week later I received a letter saying my request had been approved as a one off exception.

The conference was amazing! Everyone was so welcoming. I remember when we arrived, a lady came up to us and said "hello" and told us she arrived yesterday and that the room was nice and the breakfast was good. She was lovely, but the funny thing was we couldn't remember who she was. That's the thing, I have met so many amazing people on the Cystinosis family on Facebook, but you don't really get to know their faces from a computer.

We went to the Welcome reception the next day, which is always a little awkward when you don't know anyone in a large room, but I soon found myself relaxed and chatting with everyone in no time. Even

# The Cystinosis Foundation An Agent of Social Change

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February 5, 1999

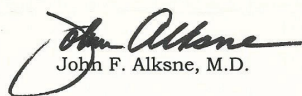
Ms. Jean Hotz  
President  
Cystinosis Foundation, Inc.  
1314 E. Santa Ana  
Fresno, California 93704

Dear Ms. Hotz:

On behalf of the UCSD School of Medicine, I am writing to express my deepest appreciation to the Cystinosis Foundation for over 14 years' dedicated support of Dr. Jerry Schneider and our cystinosis research and patient care program. I am overwhelmed to learn from Karen Stephens that due to your leadership, the Foundation has contributed more than \$380,000 during this time. The Cystinosis Foundation has surely contributed greatly to developing lifesaving treatments and to providing an improved quality of life for patients and their families as they battle this debilitating malady.

Again, please accept our warmest thanks and profound gratitude.

Sincerely,

  
John F. Alksne, M.D.

the people who could not talk the same language had the chance to communicate with each other through interpreters. This was truly special. This is one example of how this conference really does go beyond borders.

The presentations were informative and I always learn something new. We learnt about a new pro drug trial and other treatments being researched and developed which always makes me hopeful for the future. Before I went to conferences I didn't realise that there was any treatment for nausea or that other patients with cystinosis suffered with migraines like me.

Some of the seminars were a little hard

to understand but there are many slide shows and pictures to help. You get the chance to ask questions of the experts at the end. There is also an agenda book, which has some of the information written in and a notebook and pen to take notes. The doctors join the welcome drinks, coffee breaks, lunches and the Gala dinner, so you are able to chat with them then.

You can choose which discussions and seminars you would like to attend. We chose not to attend the paediatric ones and used this time to visit with others, as I am past that stage. There was also a parents' panel and an adult patients panel, which I

[SEE SEMINARS page 15]

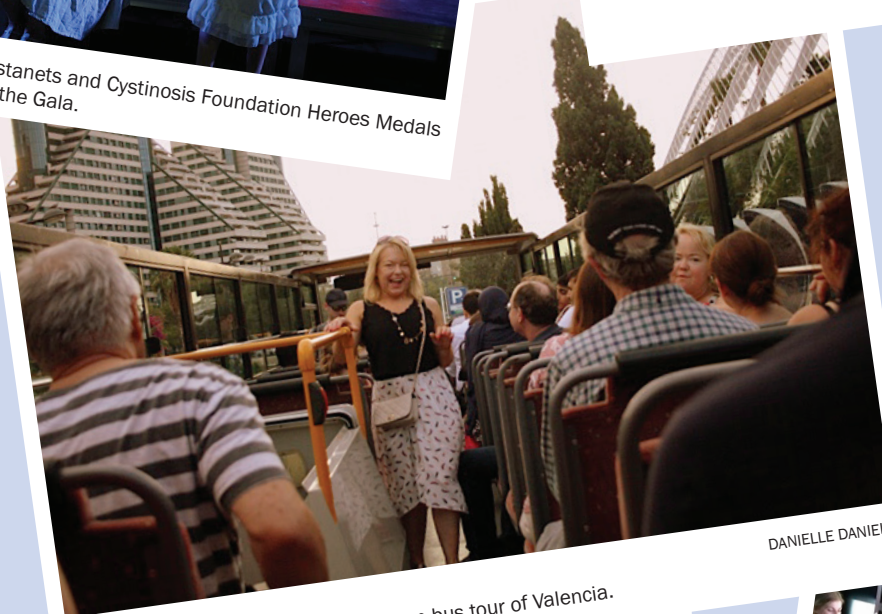


A gift of Spanish castanets and Cystinosis Foundation Heroes Medals were handed out at the Gala.

## What people say about the International Cystinosis Congress-Beyond Borders:

"Our daughter had so much fun and enjoyed the time with Don Hammond and the group. She has learned so much, but most importantly, somehow, she has gained confidence in knowing that she is not alone and was very encouraged by the young adults and their stories."

Parent of Cystinosis Patient  
United Kingdom



Ami Froelich hops on board for the open bus tour of Valencia.

DANIELLE DANIELS



Left to Right: A. Froelich, M. Drury, D. Daniels, F. Wonnink, M. Tromp, V. Gomez and J. Visser share their experiences and answer questions from the audience.



Valerie Hotz, executive director of the Cystinosis Foundation and Alexey Tsygin, MD.



Irati Vilarino Uriate and Danielle Daniels.

GAIL DANIELS





The 2016 Cystinosis Family Quilt was created by families during the conference.



MATTHEW MENCE  
Peter Bourquin and Michaela Drury during the first Family Constellation session.



Rezan Topaloglu, MD presented on care of cystinosis patients in Turkey.



Left to Right: F. Wonnink, G. Fogliada, D. Daniels, G. Daniels, M. Bos, J. Tromp and S. Goulsbra.



Physicians and patients celebrate, posing in front of the 2016 Family Quilt.



VALERIE HOTZ  
A Tiempo held everyone spellbound with their amazing Flamenco dance performance.

## ◆ NINTH from page 1

Ariceta, MD, Elena Levchenko, MD and Jennifer Simpson, MD. Their leadership contributed to making this an excellent and memorable event for everyone who traveled a great distance to learn more about cystinosis and to simply be together.

Individuals representing 45 families from 14 different countries were in attendance. Healthcare professionals from 17 nations including the United States, Canada, and countries throughout Europe and the Middle East were in attendance to share their expertise about cystinosis and learn more about this ultra-rare metabolic condition.

A lot of information was presented and also fun activities were available for all. The theme “Celebrating the Lives of Our Cystinosis Families” was apparent when we viewed the Cystinosis Family Quilt that was created in 1996. As I inspected the quilt both parents and patients walked by exclaiming, “There I am!” and pointed to their photograph on the quilt. In keeping with tradition, the conference kicked off on Thursday evening with a Welcome reception. It was nice to reacquaint with longtime pals from over the years and make new friends. Some adults enjoyed the terrace bar afterwards or ventured into Valencia to explore.

The conference began Friday morning with opening remarks by Dr. Ariceta, Lourdes Sanz and Valerie Hotz. Freek Wonnink was presented the Hobbs Humanitarian Award. After working in local government, Freek established his own software business. He has gone the extra step over the years by participating in medical studies for the purpose of learning more about cystinosis. Throughout his teens and early twenties, Freek did not walk and was confined to a wheelchair. After his physician explained there was no reason he could not walk, Freek began the very long, very difficult process of physical therapy to regain mobility and eventually met with success. His leadership and contributions to our community are appreciated.

The additional recipient of the Hobbs Humanitarian Award this year was Joshua Hotz. I had the pleasure of presenting the award to Joshua, recognizing his contributions to advancing scientific understanding of cystinosis by participating in medical

studies since his diagnosis in 1982 (when there was no treatment) often at great discomfort, in order to increase knowledge about cystinosis. Today patients benefit from the selfless contributions of Joshua and Freek.

The first session of the program was called Cystinosis Family Constellation, presented by Peter Bourquin, a therapist who lives in Barcelona. Peter specializes in Family Constellation Therapy, a therapeutic method which draws on family systems therapy. He started off with a group meditation exercise, then moved into three separate sections inviting volunteers from the audience. Peter speaks Spanish, German, Italian and English and was excellent at translating participants’ comments into English as he extemporaneously guided discussion. At the same time, simultaneous translation was available for participants using the headsets.

The first session on Friday focused on a person living with cystinosis. With guided questions he had this person think about the illness, accepting her destiny, her body and her health. Also discussed was how the condition impacts personal relationships.

The second session focused on parents of a patient. Among the topics open for discussion was how the condition impacts their marriage over time. The third constellation was with a sibling of an affected brother or sister. These were very thought provoking and definitely emotional sessions that the participants enjoyed and appreciated. Families and individuals from Italy, Spain, Ireland, the United States and the United Kingdom participated in and observed these sessions.

After lunch the plenary session began with Dr. Gahl speaking on the Genetics of Cystinosis, Dr. Goodyear presented on Managing Fanconi Syndrome, and Dr. Grimm gave a talk about Renal Transplantation, from A to Z. We then had the first mini-symposium, Be Strong - Development of Muscle Mass and Bone Density in Cystinosis. Medical experts from several different countries presented on aspects of this topic.

The first presentation was Dr. Doris Trauner, discussing Muscle Wasting in Cystinosis, followed by Dr. Ranjan Dohil, who presented on Feeding in Cystinosis. Dr. Mary Leonard delivered her presentation on Impact of Cystinosis on Bone

and Muscle Development, an area she and Dr. Grimm are researching. Children and adults with cystinosis have multiple risk factors for fragile bones, including loss of phosphorus in the urine, abnormal Vitamin D metabolism, chronic kidney disease, thyroid disease, low muscle strength and decreased weight bearing physical activity.

An excellent panel discussion featured parents answering questions from other parents including the parents of younger patients. Not only was this helpful for other parents to learn from those with more years of experience, the healthcare professionals also appreciated this program to understand issues from a parent’s perspective.

A new feature of the conference program involved a panel of nine medical experts representing nine different countries who addressed the topic, Rare Disease Multi-specialty Clinics; how it is done at my center. It was illuminating to see the variations of how cystinosis clinics are operated.

At the conclusion of Friday’s program, guests who signed up in advance enjoyed a wonderful guided tour of the city of Valencia. We rode on an open roof double decker bus and ended up in the older part of the city, where we hopped off and shared dinner with new friends we had just met that day.

A very valuable and informative patient panel discussion took place Saturday morning. Danielle Daniels, Michaela Drury, Ami Froehlich, Victor Gomez, Mark Tromp, Jorin Visser and Freek Wonnink spoke about their journeys, what is important to them and answered questions from the audience.

There were more mini-symposiums, which was an effective way to organize the volume of complex material for families. The first was See the World - Cystinosis and the Visual System led by Dr. Jennifer Simpson from the University of California at Irvine. Many issues about the eye were discussed by various doctors from different countries. A helpful parallel workshop focused on the most efficient technique for administering eye drops so that none of the medicine is wasted.

Dr. Nieves Martin-Begue presented on Intracranial hypertension and Nephropathic Cystinosis, something all patients

[SEE TALKS page 15]



# ◆ BONE from page 4

fessor and Chairman in the department of Pediatric Kidney, Liver and Metabolic Diseases at Hannover Medical School in Hannover, Germany, Dr. Dirk Schnabel, Deputy Head of SPZ for chronically ill children and adolescents at the University Medicine Berlin in Berlin, Germany and William A. Gahl, MD, PhD, Clinical Director, at the National Institutes of Health National Human Genome Research Institute (NHGRI) and Director at the Undiagnosed Diseases Program in Bethesda, Maryland, USA.

*“It is encouraging news that cystinosis experts from many different nations are gathering to address this pressing issue.”*

– Valerie Hotz

Other participating experts include, Frank Rauch, MD, Professor of Pediatrics, McGill University, Shriners Hospital for Children in Montreal, Canada, Aude Servais, MD, Service de Néphrologie adulte at Hôpital Necker, Gema Ariceta, MD, Head of Nefrología Pediátrica at Hospital Vall d' Hebron in Barcelona, Spain, Dr. Atif Awan, Consultant Paediatric Nephrologist at The Children's University Hospital in Dublin, Ireland, and many other experts will be participating.

Valerie Hotz, Executive Director of the Cystinosis Foundation, will attend the meeting and report the outcome. “It is encouraging news that cystinosis experts from many different nations are gathering to address this pressing issue. We believe bone and muscle development is of paramount importance to everyone - children and adults alike - and is one of the most serious complications for an adult Cystinosis patient. We need to be educated on this topic, especially if there are preventative steps that can be taken to lay a foundation during youth for stronger bones and muscles for adult patients,” says Hotz.

“We have a medical writer available for this conference and our goal is to complete the paper on the outcome of this meeting quickly,” says Hohenfellner.







In 1985 Bruce Mund, member of the Cystinosis Foundation Board of Directors, presents the first check ever from a patient support group for cystinosis research to Dr. Jerry A. Schneider.



Left to Right: Daniel Kelly and Maria Pekli at our 2010 conference in Italy.



Riccardo Fogliada and his mother Mara Mazzina listen to presentations at our conference in Italy, in 2010. Mara is the president of the support group in Italy and helped organize and host the program.



Left to right: Dr. Jess Thoene, Cystinosis Foundation President Jean Hobbs Hotz, Peter and Lesley Greene, Founders of the UK charity, Children Living with Inherited Metabolic Diseases (CLIMB), at the NORD Conference in 1995.

# Faces in the Crowd

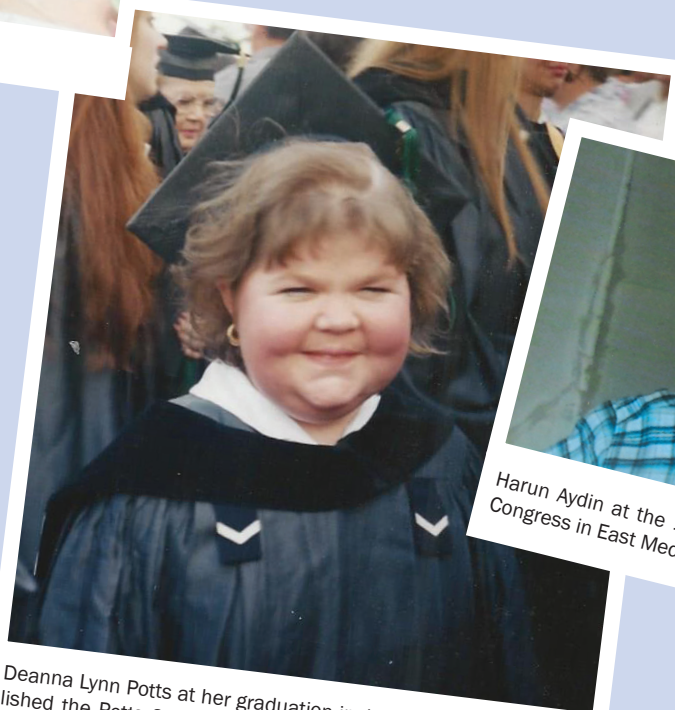




Betul Eroglu enjoying our conference in Ankara, Turkey, 2009.



Founding Cystinosis Foundation board members Wendy Clark and Sue Russell hosted the first ever gathering of families with Jerry A. Schneider, MD in attendance, in Oakland, California in 1983.



Deanna Lynn Potts at her graduation in 1995. Her parents established the Potts Scholarship in her memory, awarding a \$1,000 college scholarship to a Cystinosis patient annually.



Harun Aydin at the 1st International Cystinosis Congress in East Mediterranean, October 2009.



Left to right: Holly Reuter, member of the Cystinosis Foundation Board of Directors, and Don Hammond share a laugh at the 6th International Cystinosis Congress, 2010, in Lignano Sabbadoro, Italy.

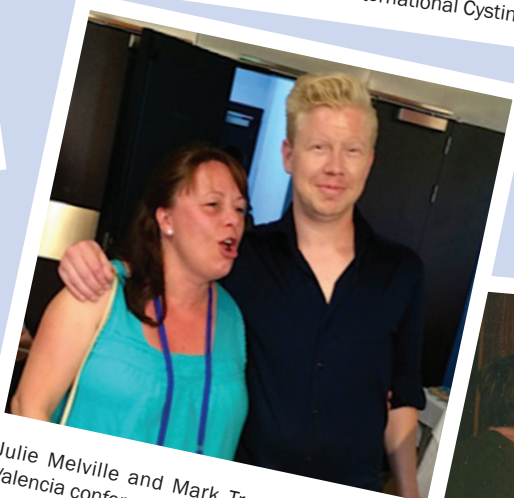




FRANK RITCHIE  
Francesco Emma, MD, served as scientific advisor at the 2010 congress in Lignano Sabbiadoro, Italy.



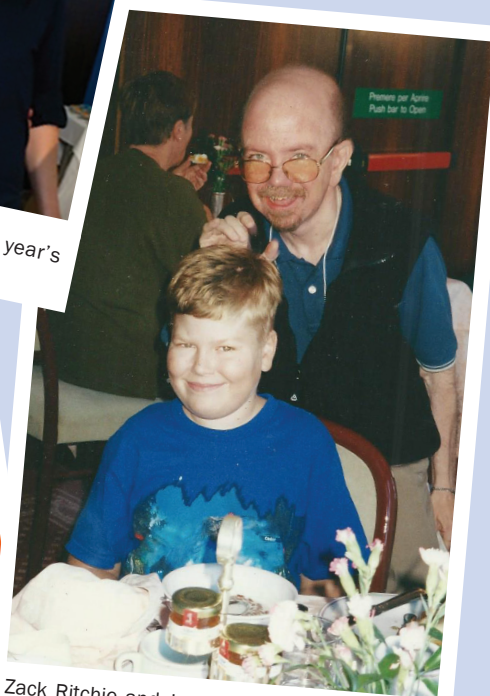
Jan, Antje and Lea Sgundek at the 8th International Cystinosis Congress in Manchester, 2014



Julie Melville and Mark Tromp at this year's Valencia conference.



After all that dancing it was time for a rest!



Zack Ritchie and Jonathan Terry at our conference in Bergamo, Italy in 2000. Happy Birthday Jonathan!



## ◆ TALKS from page 10

and parents should be knowledgeable about and Dr. Doris Trauner presented a talk on Visual Spatial Deficit in Cystinosis and school related challenges. While not every patient has these challenges, it is important to be informed and request special learning accommodations from your child's school if your child is in need of these services.

The mini-symposium, Learn and Love – Transitioning to Adulthood in the Face of Cystinosis was moderated by Dr. Elena Levchenko from the Netherlands. Dr. Levchenko reported happy news during her talk, Male Fertility. A male Cystinosis patient has successfully fathered twins. Previously it was believed that males treated with cysteamine are not fertile. Dr. Levchenko discussed the possibilities of in-vitro fertilization resulting in pregnancy and reported more research is required to understand mechanisms of male infertility in Cystinosis.

Dr. Bertholet-Thomas delivered a presentation on Endocrine system in Cystinosis, and Medication Compliance Issues was addressed by Dr. Ariceta. Dr. Servais reported in her talk, Care of Adult Cystinosis Patients, over 50% of patients liv-



Merle Mund, Cystinosis Foundation Director gives awards out at the Gala. With their own daughter Krista having passed away last year, Merle and her husband Bruce continue to support our community.

ing with cystinosis are now adults. While most patients who have been treated regularly since the time of diagnosis with cysteamine have been shown to develop renal failure later in life, most patients ultimately require a kidney transplant. It is clear there is need for both increased awareness of the disease as well of how it is managed for adults.

The final mini-symposium was called The Future is Bright – Novel Drugs, Diagnostics and Therapeutics and was moderated by Dr. Ariceta. This session featured Dr. Dohil's Update on Delayed Release Cysteamine. This treatment has been approved for use in the United States and in Europe, with reimbursement approved

[SEE GALA page 16]

## ◆ SEMINARS from page 7

hosted with several others. We answered questions about how we deal with cystinosis. For example I talked about things I have had to overcome in my life and how my transition from paediatrics to adult care went.

Childcare was provided and they even went on a day trip to the Oceanarium. We had a really fun session called laughter therapy. This was slightly awkward to start with, but was truly as funny as it sounds and very helpful. I went for a meal with some other cystinosis adults, and thoroughly enjoyed a bus tour of Valencia that was prearranged by the Cystinosis Foundation.

On the last day we had a huge buffet and amazing Flamenco dancers and music. I normally struggle with social life, but I really felt part of this wonderful community and was able to open myself up and enjoy the company of everyone who is a part of it.

## ◆ PRESIDENT from page 2

us, we look to forward to a brighter future, one that includes the development of improved treatments for cystinosis patients. This will happen because of the contributions of patients and scientists pursuing promising research. Scientists bring truth to the equation and patients bring the spirit. I share Dr. Schweitzer's sentiment when he observed, "Because I have confidence in the power of truth, and of the spirit, I have confidence in the future of mankind." I invite you to join me in sharing Dr. Schweitzer's confidence in the future of all cystinosis patients and of all mankind.

With my warmest wishes to you and your loved ones for the happiest of holidays and a joyous New Year, I am grateful for your being part of this incredible global community.

With warmest regards,  
Jean Hobbs-Hotz  
Founder and President

*I don't know what your destiny will be, but one thing I do know: the only ones among you who will be really happy are those who have sought and found how to serve.*

– Albert Schweitzer

## ◆ HAMMOND from page 5

as well as our shared destiny. We give thanks to those who made it possible for us to be here today and contemplate the path before us. As we set our eyes on the path ahead, we understand it will include breakthroughs, it will include sorrow, it will include healing ourselves and it can enrich our lives and draw us together. But only if we allow it.

Eleonor Roosevelt once said, "The future belongs to those who believe in the beauty of their dreams." We are the cystinosis community and we must embrace our dreams that one day our loved ones will be healed.

# In Loving Memory of Annie Fencel

By Tom Fencel

Andrea Fencel - known as Annie to all her friends - was born the third of three children. At the early age of nine months she was diagnosed with cystinosis. At the time she was one of the youngest people to be diagnosed with the condition. As you might expect, most of the medical community here in Omaha had never even heard of it.

Like typical cystinosis patients, it was necessary for Annie to travel to many different places to see many different doctors. Because of that we scheduled most of our vacations around the destinations of Annie's medical appointments and Cystinosis Foundation conferences.

Annie was one of the experimental patients for what would be Cystagon. The first few days of the treatment were terrible but she got used to it, quite an accomplishment for a one year-old. Outside of her constantly having to take medication and the occasional throwing up, Annie's life was much the same as any other kid her age.

Although she was small she loved to dance, and she loved school (not so much for the learning, more for the friends). Her childhood went on into adolescence normally but her kidneys were beginning to suffer the consequences of her disease.

At 15 she had to go on dialysis and we were sent to Stanford University to see about a transplant. Here we met the people that we all agreed were the best doctors

we had ever met. Annie's second cousin Dean Prohaska gave her a new lease on life with the greatest gift ever, a kidney.

Shortly after the surgery Annie was back home and running in high gear. She resumed dance, school, work and fun with her friends. In one of her return check-ups at Stanford she was invited to attend "Transplant Camp" at St. Dorothy's Rest near Santa Rosa, California. After attending one time it became her home away from home and still is to this day. Unlike most of her friends back home all of her friends at St. Dorothy's were other kids who had to face huge medical issues. She loved it so much there that when she became too old to be a camper she returned as a camp counselor.

Annie enjoyed a variety of accomplishments. She made the honor roll at school. Her dance teams won numerous awards. She graduated from high school on time even though she missed a large part of her Junior year. She participated in many plays and stage productions, even choreographing one while she was on her second round of dialysis. Annie spent some of her free time helping young adults with special needs. She even made her own short documentary about living with a transplant called "A Day In The Life", which you may view online at <https://www.youtube.com/watch?v=kWshA8qtia8&sns=fb>.

Like all young people sometimes we do stupid things. Annie was no exception. Not taking her anti-rejection medica-



Annie Fencel

tion (this is a strong hint to anyone with a transplant) caused Annie's kidney to eventually fail and put her back on dialysis. After struggling for two years Annie's body finally had enough and she passed away.

Annie had many friends and family all over the country. She treated all of them as her best friend because to her they ALL were. Even after a year and half of her being gone the people she called her "best friends" still continue to message her on Facebook.

Annie's sister Brooke probably described her the most appropriately when she sang the song "Astonishing" at her funeral. Annie packed 75 years of living into the short 26 years she was given to us and then God recruited a very special angel. "Smile A While".

## ◆ GALA from page 15

only in the United States and Germany. Dr. Emma explained his ongoing research of the current approved drugs with the goal of determining if there is any other approved medication that could effectively treat cystinosis.

The Saturday evening Gala buffet dinner featured delicious Spanish dishes and a fabulous Flamenco dance show, culminating in the children giving an impromptu dance show of their own to everyone's delight. I thoroughly enjoyed delivering a gift of castanets to every patient along with our own Cystinosis Foundation Heroes Medal.

Throughout the conference people were encouraged to decorate their own square of fabric. These were then sewn into a lovely new Cystinosis Family Quilt 2016 and was unveiled at the Gala dinner. There was a silent auction with lovely glass items donated by Lee Knaus and watches donated by Xavier Wehe. Lee also donated glass beads for the children to make bracelets, anklets or necklaces and the children were wearing their jewelry creations at the Gala dinner.

While the teens enjoyed just being together, they also experienced an exotic afternoon field trip to the L'Oceanografico, the largest aquarium in Europe, a short

walk from the Barcelo Hotel.

Don Hammond was not able to attend the conference but he did a wonderful PowerPoint presentation honoring our real heroes, the trailblazing patients who have passed on, as well as those who are with us today. Our heroes continue to contribute to their communities and help others with this condition.

Dr. Jerry Schneider wrote a letter honoring our late daughter, Krista Mund, which was read by Dr. Paul Grimm. Dr. Schneider recognized her as being a true pioneer in the beginning of the use of cysteamine. They are all our heroes and they shine brightly.



# In Loving Memory of David Reuter

By Holly Reuter

On June 14, 1992 David Daniel Reuter was born prematurely with a birth defect. His intestines were on the outside and we had to rush him by ambulance to the University of Minnesota Neonatal Care Unit for a Gastroschisis repair surgery. This would be the first of a series of medical interventions that he would need throughout his life.

He was very small - 5 pounds, 4 ounces - when he was released from the hospital. In the next months of his life, we didn't realize that all of the vomiting, extreme thirst, photophobia and failure to thrive were due to an ultra rare genetic disease called cystinosis. At his 18-month check-up, the lab work revealed that he had protein and glucose in his urine.

We took David to the ophthalmologist to check his eyes for cysteine crystals. The doctor had to look in a big medical book to see what he was looking for. That finally gave us an answer to what was going on with David. They explained about cystinosis and told us there was an experimental drug we could give him called cysteamine. It smelled like a combination of sulphur and rotten eggs.

The prognosis was that some children lived to be eight or ten years of age and then went into renal failure, but that the experimental medicine could delay this. David started a regimen of taking this experimental medicine every six hours and the additional supplemental medicines.

David was very smart and liked science. He did well at the science fair in elementary and high school, winning ribbons in both. He memorized jokes, and the nurse at school told us that David would tell her a joke every day. For example: Why did the golfer wear 2 pairs of pants? In case he got a hole in one. He loved Weird Al Yankowitz and other comedians.

David and his dad went to Tae Kwon Do and earned their yellow belt-green stripe. He started drawing comic strips with animals. One of these he made up using our dog and cat called "The Adventures of Bengal and Magic." He got a few of these printed in the Lake Region Times, our local paper. He was active in Boy Scouts throughout high school.

David was very determined. On a Boy



David Reuter

Scout high adventure trip to Montana, the scouts were climbing a mountain up to a glacier lake. His health was already somewhat compromised and his dad, Dan, admitted that he would probably have turned around without getting to the top. They were the last ones and were worried

that they would miss the boat. David was determined that they would reach the top and according to Dan, it was worth it. David completed all of his merit badges and did a pancake breakfast fundraiser for his Eagle Scout project. Then, we took the money and packed meals for Kids Against Hunger. David went deer hunting with his dad and also became scuba certified and enjoyed diving a few times in Mexico.

David played the trumpet in band until it also became too difficult. He was in Knowledge Bowl and participated in Drama, one of his favorite activities in high school. In the summers, David attended Michelangelo Art Camp at Bethany Lutheran College. This was the first time that David painted with oils. He continued to take art classes in high school and later in college.

David also enjoyed playing chess and participated in some chess tournaments even winning at times. As a family this cystinosis journey led us on many vaca-

[SEE REUTER page 18]



Minnesota Spring, by David Reuter.

# In Loving Memory of Brian Kent Riddle

**March 22, 1976 - February 14, 2016**

By Barbara Riddle

*I have fought the good fight, I have finished the race, I have kept the faith.*  
2 Timothy 4:7

This scripture reference sums up another life lost in a 39-year battle with cystinosis, end-stage renal disease, and encapsulating peritoneal sclerosis, a life-altering complication of peritoneal dialysis. My son Brian Riddle was one of what I would call the “old guard” - patients participating in early National Institutes of Health (NIH) trials of cysteamine therapy. We were thankfully exposed to the latest research being coordinated by the NIH and other entities under the direction of such pioneers as Dr. Joseph Shulman, Dr. William A. Gahl, and others.

Brian’s life was one of resilience and a spirit driven to success in spite of his illnesses and the myriad of complications encountered along the way. Brian received three kidney transplants before the age of 16. His father and I were among his donors. After losing three transplants to rejection, he began what I call his “career on dialysis.” His first option was peritoneal dialysis (PD), which lasted for about 11 years.

Instead of succumbing to self-pity, Brian finished high school as an honor roll student in spite of being homebound for a time. He attended a local community college for a year to reset his ambitions, worked three part-time jobs, and moved on to the University of Pittsburgh, graduating with a Bachelor’s Degree in Information Technology. Brian accomplished all of this while doing peritoneal dialysis. And yes, he had time for participation in paintball tournaments. He and his brother also started a deck hockey team, which competed in local leagues.

Upon graduation, Brian secured employment with Highmark Administrators in Pittsburgh. He bought a home. After recovery from bilateral quadriceps ruptures in 2004, Brian was stricken with his greatest challenge in April of 2005, encapsulating peritoneal sclerosis. This often fatal complication of PD caused him to reluctantly leave his career and move to Virginia with us, as we were connected to



Very few people know how medically challenged Brian was in his final days, but he never complained about his mortal situation.

home hemodialysis.

Brian’s relocation by no means led him to give up. Via the Internet Brian was able to connect with a dialysis patient from Chicago who initiated a dialysis group. Brian used his computer skills to create a web page, and together he and his cohort began a group, NxStage Users, which advocated home hemodialysis. The group still exists today under the name of Home Dialyzers United (HDU) and is a 501(c)(3) organization.

They organized the first ever meetup conference of dialysis patients from across the United States in Las Vegas in 2010. It brought dialysis users of the NxStage System One together along with doctors, nurses, vendors, and advocates within the dialysis community. The goal was to inform patients of the home dialysis choices that are not often given at the time of transition to dialysis.

Brian left his position as Vice President of the group in 2012, as his health began to decline with frequent small bowel obstructions and abdominal abscesses. He wasn’t finished. He began a thriving Facebook page, Dialysis Discussion Uncensored, in May of 2012. We now have over 10,000 member participants of this vibrant group. Brian continued to offer support within his group and behind the scenes, ever so humbly until his health took a fatal turn.

Brian passed away on Valentine’s Day

of 2016. His father and I were at his side, holding his hands as he passed from life to death. Brian suffered greatly in the last months of his life. He requested that the group, which is now his legacy, would continue as an open public forum, allowing the dissemination of information about renal disease and its challenges. Patients don’t always know they can live longer with better lives on home dialysis modalities.

Brian’s brother summed up his life best when he said, “Brian stoically taught the power of perseverance and the hidden strength within meekness.”

## ◆ REUTER from page 17

tions. Attending Cystinosis Foundation family conferences, we visited California, Michigan, New Hampshire, Nevada, Illinois and many places in between.

This became our extended family-this extended cystinosis family. We are especially thankful for the Cystinosis Foundation, our Midwest group and their support. After graduating from St. Clair High School in 2010 David underwent kidney transplantation and received a new kidney from me, his mother. He made the transition from taking electrolyte supplements to immunosuppressant therapy.

In the fall of 2010, we flew to Italy for the 5th International Cystinosis Confer-

[SEE DAVID page 19]



## ◆ DAVID from page 18

ence, hosted by the Cystinosis Foundation. We visited Venice, Florence and Rome. It was fantastic. Italy has such great artwork and we were able to see a lot such as Michelangelo's "David," the Sistine Chapel and the Leonardo Di Vinci Museum.

David moved into the dorms at Bethany Lutheran College and continued to study art. David's muscle weakness continued to worsen and he started getting all of his nutrition through a g-tube because swallowing food put him in a choking risk and also risk of pneumonia due to aspiration. He had severe muscle weakness in his hands and had a hard time opening bottles, etc. We found some aids to assist with this or sometimes David would just leave the bottles open.

Through all of this, David never complained. He would ask for things or for help with something that he couldn't do on his own, but never complained about everything that he had to do or all of the things that he couldn't do or that he needed help to do. He graduated from Bethany Lutheran College in December 2014 with a degree in Studio Art.

David concentrated on mostly oil and acrylic paintings or drawings. For his senior project, he painted mostly abstracts. He found this form to be enjoyable since it was not making an exact duplicate of a photograph, but a free form to do whatever his wanted. He won a prize for his abstract "Minnesota Spring." It has green with colorful spots and white on top because it depicts the grass and flowers that bloomed, but the snow fell over the top.

After graduation David moved home and we made plans for his next two surgeries. His muscle weakness had compromised his posture and he had both severe scoliosis and kaiphosis. We made plans for him to be in Halo traction and then have a spinal fusion surgery after 4 weeks of traction. Again, David went through all of the appointments and tests prior to the surgeries without complaint. The Halo was less than comfortable, but David just put up with it even though he didn't like it. It was amazing the difference that it made. His spine had really straightened out and we all commented on how his speech was better since his posture was better.

On Monday, August 17th, David had his 13th operation. We spent time together

# In Loving Memory of Krista Mund

By Merle Mund

Krista Noelle Mund was a kind soul with a strong spirit. She always hoped for the best, but if things went wrong her response was always, "Okay." Her strength and spirit were always directed toward making people she loved and cared about to feel special and accepted. Krista loved to laugh and shared stories with her friends and sometimes she would surprise you with some "sassy" drama and she loved that too.

Krista was a very simple person. She loved to go out to eat with friends, sit with the dogs at home or spend time on the Internet, interacting and planning things with people. She had an infectious laugh and a lively, genuine smile that stood out in any room. Krista was special in many, many ways. Although she had many health complications, she always chose to shine bright and push forward, looking ahead.

She contended with and fought tirelessly against the many problems of Nephropathic Cystinosis her entire life. At the age of eight in 1978 she volunteered to be the sole medical research subject at University of California at San Diego with Dr. Jerry

Schneider. She remained in the hospital for nine weeks in order for physicians to determine the safe and adequate dose of cysteamine for cystinosis patients.

Eventually this drug became the sole

in the hospital, prior to his last surgery, playing cards. David and Shawn played chess together. The spinal fusion took 12 hours to complete since they were making an incision from the front and also from the back. Everything was going well. On Wednesday, he was determined to get out of bed. He sat in a chair and moved around the room a little. David remained insistent about walking. He convinced the nurses to let him walk out of the Intensive Care Unit, through the hospital and out to the skywalk so that he could see outside.

It was wonderful to hear he had such a good day. During the night, David tried to cough up some secretions and struggled. At 4:00 a.m. they tried to help him with cough assist, but it was not working. They put his breathing tube back in. Every-



Krista Mund

oral treatment for Nephropathic Cystinosis and was manufactured in capsule form as Cystagon by Mylan in 1994. Raptor Pharmaceuticals conducted clinical trials for an enteric-coated delayed-release form of oral cysteamine. It is now manufactured by Horizon Pharmaceuticals and is available as Procysbi. This therapy requires dosing every 12 hours. These two treatments, Cystagon and Procysbi, are currently the sole oral treatments for the disease of cystinosis and are prescribed throughout the world for people afflicted with cystinosis.

Krista passed away October 20, 2015 at the age of 45.

thing seemed fine, but then his heart rate dropped and he went into cardiac arrest. I got a phone call at 5:34 a.m. to explain that he had a pulmonary embolism. We started to get ready to drive to the hospital, but at 6:01 a.m. we received another phone call that David was getting weaker and he would not make it until we got there.

We said our goodbyes over the phone. On August 20, 2015 after 23 years of taking horrible tasting meds at 6:00 a.m., 9:00 a.m.; noon; 3:00 p.m.; 6:00 p.m.; 9:00 p.m. and midnight, doctor appointments, blood draws, ultrasounds, EKGs, and much more, David is finally at peace. We will remember him for his determination, courage, strength, humor and creative spirit.

# DONATIONS

Anthony Alvarez  
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Michael Vellard, Ph.D.  
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Frederic Wilson

## MEMORIALS

IN MEMORY OF  
BARBARA BROCE  
Nelda and Ed Bender  
Kelly McGuire  
V. J. P. Farms

IN MEMORY OF ANDREA  
FENCL  
Martha Abbott-Magnuson  
James Abel, D.C.  
Kathleen and Lee Arends  
Rebecca and Stephen Baker  
Clarice and Frederick Barnhill  
Terri and Calvin Bull  
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Dr. Paul and Luann C. Dritley  
Elkhorn Band Boosters  
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Mary and Douglas Fencel  
Ruby Fencel  
Teresa and Shawn Fernald  
Michael D. Guillatt CFP  
Verna and Larry Headley  
Carmen and John Housley  
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Karen and John Jorgensen  
Theresa Kreifels  
Nancy Neumeyer  
Kenneth I. Peterson  
Rose Marie Rosengren  
Lori and Neil Sergel  
Michele and Marty Thompson  
Mary and John Thomsen  
Lori Jane Wildhagen  
Houghton Vandenack Williams

IN MEMORY OF  
WILLIAM KNODELL  
Susan and Brooke Taney

IN MEMORY OF KRISTA  
MUND  
Winona and John Avila  
Paul Baty  
Sally and Bill Bjorge  
Gayle and Ernest Britt  
Mary Lou Brooks Trust  
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Marilynne and Neil A. Elverson  
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Joyce and Nori Umemoto  
George Walker

IN MEMORY OF DEANNA  
LYNN POTTS  
Gail Potts

IN MEMORY OF DAVID  
REUTER  
The Alitzer Family  
JoDell Anderson  
Rebecca Carpenter  
Melanie and Cliff Colby  
Julie Fitzsimmons  
Leah Frank  
Carol Frederickson

Thank you to all who have donated to the Cystinosis Foundation. We are grateful.

## Your Gift Saves Lives and Connects Families

By Valerie Hotz

When you donate to the Cystinosis Foundation, you support life-saving programs that include our educational family conferences, effective advocacy, research, improving patient care through educational programs for healthcare professionals and reaching new patients and families who are at risk.

Our mission is global in its reach and impact. In addition to helping families who are isolated to join our international community, your gift contributes to

strengthening the scientific network, leading to further collaborations that ultimately benefit patients. The more physicians we educate about Cystinosis symptoms, the sooner young patients may be diagnosed and begin crucial treatments and the sooner they begin enjoying an improved quality of life.

The late highly esteemed scholar, Joseph Campbell, famously observed, "Money is congealed energy and releasing it releases life's possibilities. In the living of life today, money is a facilitating energy source.

With money in the tank like gasoline, you can get places you otherwise couldn't go."

Your donation releases life's possibilities. By giving to the Cystinosis Foundation, you help us to accomplish our mission and you help fight cystinosis. Every gift makes a difference. We would not be where we are today without the generous financial support of our caring families and friends.

We gratefully acknowledge the loyal and continuing support of those who have

[SEE GIFT page 21]



# TIONS

Lori Ganske  
Kathleen and Frank C.  
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Mildred and Elwin Gunnink  
Sandra Gunnink  
James Hannaman  
Noreen Hannaman  
Maureen Hollerich  
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Sandy Wersal  
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Sue Wilde

Kelly and Troy Zabel  
  
IN MEMORY OF  
MASON REED  
Kelly McGuire  
  
IN MEMORY OF BRIAN  
RIDDLE  
Annette and Carl D. Bradley  
Mike Bradley  
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BRINKS  
Mary Joan Goessling

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PEACHMAN  
Gary Dilillo

IN-KIND GIFTS  
Thomas R. Brown  
Excellence in Pediatrics  
Institute  
Lee Knaus  
Merle and Bruce Mund  
Jamie Westdal Photography  
Orphan Europe Recordati  
Group  
Serena Scott  
Susan Scott  
Xavier Wehe

GRANTS  
Orphan-Europe Recordati  
Group  
Raptor Pharmaceuticals  
Sigma-Tau Pharmaceuticals

have made a gift to the  
ateful for your financial support.

### ◆ GIFT from page 20

directed their philanthropy to the Cystinosis Foundation. They have provided the gasoline in our tank to help us improve life for thousands of Cystinosis patients and their families since 1983. Historically, together we have helped keep research alive when other funding sources disappeared.

We encourage you to consider making a gift to the Cystinosis Foundation today. Make your contribution online via secure PayPal processing or by Visa, MasterCard, American Express or Discover card, or by sending your check to the address

on the membership application. Visit our web site at [www.cystinosisfoundation.org](http://www.cystinosisfoundation.org) today and click on DONATE at the top of the home page. Many employers make matching gifts and yours may be one.

Support us when you shop on Amazon. Simply go to [smile.amazon.com/ch/94-2927892](http://smile.amazon.com/ch/94-2927892) and Amazon donates to Cystinosis Foundation of California Incorporated (the official name we incorporated under in 1983).

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



Support the Cystinosis Foundation's Mission when you shop on Amazon. With holiday shopping in full swing you can make a difference by making purchases at [smile.amazon.com/ch/94-2927892](http://smile.amazon.com/ch/94-2927892) and Amazon donates to the Cystinosis Foundation of California Incorporated.

# Deanna Lynn Potts Scholarship 2016

## 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 2013:

Address \_\_\_\_\_

City \_\_\_\_\_ State \_\_\_\_\_ Zip Code \_\_\_\_\_

Email \_\_\_\_\_

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

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

(Applicant Signature) \_\_\_\_\_ Date: \_\_\_\_\_

## Scholarship Criteria

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

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

### APPLICATION PROCEDURE:

Applicant must submit by April 12, 2017:

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

An official copy of high school transcript

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

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

For complete guidelines for submitting an application, visit our web site at [www.cystinosisfoundation.org](http://www.cystinosisfoundation.org)

### Cystinosis Foundation Mission

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

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



We are grateful for the extremely thoughtful and generous  
in-kind gifts from the following friends:

Thomas R. Brown  
Excellence in Pediatrics Institute  
Lee Knaus  
Merle and Bruce Mund  
Orphan-Europe Recordati Group  
Serena Scott  
Susan Scott  
Jamie Westdal Photography  
Xavier Wehe

## Cystinosis Foundation Membership Application

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

(PLEASE PRINT)

Honor Circle	20,000
Patron	10,000
Lifetime	2,500
Visionary	1,000
Professional	200
Supporter	100
Family	50
Individual	25

Yes, I want to become a member of the Cystinosis Foundation.

Enclosed are my membership dues of \$ \_\_\_\_\_

No, I do not wish to become a member at this time, but I do wish to make a contribution.

Enclosed is my gift of \$ \_\_\_\_\_

Please accept my donation of \$ \_\_\_\_\_, given

In Honor of \_\_\_\_\_

In Memory of \_\_\_\_\_

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

NAME \_\_\_\_\_

ADDRESS \_\_\_\_\_

CITY \_\_\_\_\_ STATE \_\_\_\_\_ ZIP \_\_\_\_\_

PHONE \_\_\_\_\_ E-MAIL \_\_\_\_\_

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

Name of employer \_\_\_\_\_

Do you belong to an organization that may be interested in a fundraising activity for the Cystinosis Foundation? Yes No

Please contact me to discuss planned giving options. Yes No

**You may make a gift to the Cystinosis Foundation online at [www.cystinosisfoundation.org](http://www.cystinosisfoundation.org).**



Cystinosis Foundation  
58 Miramonte Drive  
Moraga, CA 94556  
USA

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We thank our longtime industry partners for their  
continuing generous financial support of our  
mission:

Orphan-Europe Recordati Group  
Raptor Pharmaceuticals, Inc.  
Sigma-Tau Pharmaceuticals, Inc.

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

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