

March 3, 2005

Dear Family, Friends, and Colleagues:

How fortunate and blessed are you? It poses an interesting philosophical question to ponder. For the Greeley family, the answer to the question would be a resounding, "Enormously." As the 5<sup>th</sup> birthday of our son, Jack approaches this March 17, 2005, we are reminded each and every St. Patrick's Day of our good fortune and blessings because four years ago we thought that we might never see our son's 2<sup>nd</sup> birthday, let alone Number Five.

As many of you know, Jack has cystinosis (SIS-TIN-OH-SIS), a rare metabolic, genetic disease that can potentially destroy every major organ system in the body – muscular, skeletal, neurological, gastrointestinal, and cardiovascular, among others. With Jack's well-being at stake in the spring of 2001 when he was first diagnosed at one year old, we decided to support his cause through the Cystinosis Research Network (CRN), a nationally recognized 501(c) (3) advocacy group focused on improving the livelihood of those with cystinosis through more research, better treatments, increased awareness, and accessible family support, while ultimately seeking a cure. More information can be found at [www.cystinosis.org](http://www.cystinosis.org).

Our efforts as CRN board members have been multi-purposed; fortunately, we are battling cystinosis with some incredible families and have been unbelievably blessed and inspired by tremendous support and encouragement from people like you. In honor of Jack's 5<sup>th</sup> Birthday, we seek your continued support and assistance in any number of ways, whether donations, corporate support, fundraising, or volunteering.

For us, our good fortune is most apparent in the reality and horror of what cystinosis and its treatments could be. First of all, diagnosing cystinosis is very challenging because there are so few documented cases and its most obvious symptoms, mainly failure to thrive (no growth) and profuse vomiting, could be caused by so many things. It is estimated each month that passes without diagnosis and treatment could cost a child one year of renal function. The treatment for cystinosis (and Jack's main medicine) is a miracle drug called Cystagon, the brand name for cysteamine. The body is naturally supposed to remove an amino acid called cystine from each cell. Jack's body chemistry is incapable of doing that, but Cystagon performs that function. If this process does not take place, cystine crystals accumulate throughout the body, particularly in the kidneys, eyes, liver, muscles, pancreas, brain and white blood cells. Without specific treatment, children develop end stage renal failure and require kidney transplants for survival. A web of other life threatening complications results, including muscle wasting, difficulty swallowing, diabetes, blindness, hypothyroidism, decreased pulmonary function, and neurological deterioration.

Cysteamine was first extensively studied as a radio-protective agent during the Cold War in the 1950s and ultimately Cystagon was FDA approved for the treatment of cystinosis in 1994. Cystagon, however, is a very harsh medicine in smell, taste, and digestive effect. Studies have shown that 14% of cystinosis patients cannot tolerate taking the medicine. Even if they can, swallowing pills four times per day presents challenges due to muscle wasting. Cystagon treatment, coupled with a slew of other medicines, creates tremendous compliance challenges. Jack currently takes six types daily and recently added Vasotec, a drug that minimizes protein from "spilling" into his urine and helps distribute it throughout the body. In its current formulation, Cystagon must be taken diligently every six hours. Jack, for example, has a 24/7/365 schedule of 8 am, 2 pm, 8 pm, and 2 am. Additionally, there is a topical cysteamine treatment in the form of eye drops that must be administered once per hour while awake to dissolve corneal cystine crystals. Children, with parents watching over them, have the best compliance, but adolescents are a different story. Try to imagine teenagers who want to take foul medicine that disrupts their lives, burns their eyes, gives them chronic halitosis, and upsets

their stomach. The discipline of compliance is not easy. With Jack, simple math tells the story. He has taken Cystagon for four years, four times per day, which totals 5,840 times. Let's knock on wood and say that Jack lives until he is 85; he would have taken Cystagon alone 118,260 times. Of course, each time he takes Cystagon it is at least four pills worth and that dose will increase as he gets bigger. If in his lifetime he averages six pills per dose, he would have taken that one drug 709,560 times! Of course, he takes more drugs than just Cystagon. You get the unnatural picture here. What else in your life would you do more of other than breathing?

Despite the harshness of cystinosis and its treatment, Jack has continued to beat the odds by being in relatively good health, perhaps one of the healthiest of all kids with cystinosis. We remind ourselves of this good fortune regularly and it is highlighted in many ways:

- Jack was diagnosed quickly in 2001 and began treatments right away. Would that have happened for Jack if we lived somewhere other than Chicago? That path has been a blessing.
- He has not only willingly taken Cystagon psychologically and emotionally over the years, but also tolerated it physically. Jack has been a champ with taking all of his other medicines, too.
- Just recently, Jack learned to swallow pills, a major breakthrough that will enable him to lose his stomach feeding tube sooner. One of those pills is chlorophyll. While not a medicine, chlorophyll does an amazing job of doing away with body odor created by Cystagon.
- Jack has seen marked physical improvement from Cystagon, including improved body chemistry levels (checked quarterly), catch-up growth (i.e. – going from below the chart in 2002 to 90<sup>th</sup> percentile now, no need for growth hormone therapy, etc.), and kidney function remaining stable at about 70% of normal.
- In 1998, the gene that causes cystinosis was identified to have 75 mutations, so there could perhaps be a less severe form of the disease; might that be Jack and might he be more compatible with known treatments?
- The brace that Jack wears at night has noticeably straightened and strengthened his feet and legs. At night, he has also shown improvement in his ability to hold his bladder and our goal is to get rid of night diapers in the next few years.
- Jack shows no signs of rickets, plus his lungs and nervous system are fully functional.

The reality is that Jack's medical care and his acceptance of Cystagon treatments have essentially commuted his death sentence to life with a chronic disease, which is our greatest blessing. Beyond the medical end of things, we see our good fortune in so many ways through Jack's development. He is a completely normal five-year-old, just less healthy than most. Street hockey, wiffle ball, puzzles, books, computer games, play dates, Yu-Gi-Oh, and Pokemon are among his favorite pastimes. Beyond a doubt though, his favorite activity is wrestling with Dad with a preference of being "on Daddy's team" and ganging up on his eight-year-old sister, Alex.

Good fortune and blessings come in so many forms for different people. For our family, Jack's progress is foremost, but our journey in life has been touched by a wonderful outpouring of support, love, kindness, and generosity from many people. When we first wrote this letter in 2003, we were really just tossing a line in the water and hoping to raise a little money; about \$18,000 came through for CRN. Then, last year, we raised almost \$37,000 in our letter writing campaign. We chose to respond to our fate and have now become committed to this cause, but along the road of raising money and building awareness, some other amazing things happened, even mushroomed, with family, friends, neighbors, and colleagues. Many wonderful people responded to our call and collectively raised money and awareness in various ways:

- Organized a family fun day at Jack's pre-school raising over \$5,000.
- Chose CRN as a \$12,000 beneficiary of the Arthur Siegel Memorial Golf Outing.
- Selected CRN as a charitable cause with their civic and social clubs, giving thousands.

- Turned annual challah bread baking for Rosh Hashanah into a \$476 donation.
- Doubled their personal donations by corporate/employer matches.
- Tapped into corporate foundations or sources for donations.
- Forwarded the letter around the office and to family and friends.

We heard it repeatedly in 2004: “We never knew. How can we help? We want to do more. What can we do?” As parents, when you bare your soul, push pride and ego aside for the love of your child, and hear these things, you get touched...plain and simple. Some say that the Lord does not give you more than you can handle. For us, we have been blessed with fresh perspective on life and know that cystinosis is part of our mission. While we, at least partially, have some knowledge, background, willpower, skills, and a network to help make a difference at CRN for those suffering from cystinosis, we also know that part of this mission is to inspire others, whether right, wrong, or indifferent.

The truth is that no one knows what tomorrow holds. For us, our lives will always be filled with questions and clouds. Will Jack continue to tolerate his medications? Will he continue to grow and have decent kidney function? Who could be a match, if Jack needs a kidney transplant? What kind of life will Jack lead (i.e. – friends, education, career, marriage, child rearing)? How long will Jack live? The fact is that 25 million people in America alone contend with some form of a rare disease. Each and every one of us has been touched by someone or something that falls in this category. It is incumbent upon all of us to strive to make a difference or as a friend says, “Put some cookies back into the cookie jar.”

Stop and consider the notion of making a difference. How can that come about? How can a regular person really contribute? Maybe you can or maybe you cannot; it is a great unknown. Think about it though. The threat of atomic warfare during the Cold War era has...some 50 years later...helped our son and others who suffer from cystinosis because scientists studied cysteamine to treat radiation poisoning. It is far-fetched and bizarre to say the least, but that’s the way science works. It is also the way miracles work; you need to try and you need to have faith.

With cystinosis, we need to raise funds for research and build awareness through CRN. In 2004 and heading into 2005, our community can look at several milestone events, some good and some bad. We lost a nine year old. We saw numerous pre-teens get pioneering, non-steroidal kidney transplants, which will be a helpful practice to all others who might need transplants of any organ. We funded a \$112,000 research study at UCSD regarding neurological development and visual processing deficit in kids with cystinosis. (Note: Jack actually participated in this study and received an MRI, cognitive testing, and neurological exam. His results showed above average intelligence, but a variance in his brain physiology that theoretically leads to perceptual and spatial performance challenges.) CRN also made a \$100,000 grant to Tulane University to study apoptosis (or “cell death”) in the cellular lysosomal cystine transfer process. Research on apoptosis not only benefits cystinosis, but also can be applied to other areas of apoptosis functions in cancer surveillance to remove malignant cells and also to remove cells that are infected with viruses. We participated in the first-ever NIH/ORD (National Institutes of Health and Office of Rare Diseases) symposium on cystinosis, which brought together some of the most brilliant minds in medicine and science. Last, but not least, we are also planning our bi-annual CRN Medical and Family Conference to be held this summer in Salt Lake City.

This is progress, but it is not enough and we need to do more. In today’s world with so many worthwhile charitable efforts, ranging from Tsunami relief to personal family causes, we ask you, our family, friends, and colleagues, to reach out to the less fortunate and help make a difference. If not cystinosis, please help elsewhere and reward and recognize your own good fortune and blessings in the process. We hope, however, that you would consider making a donation to the Cystinosis Research Network in the honor of Jack and his 5<sup>th</sup> birthday. Any amount or other type of outreach matters and would be most appreciated. We would urge you to send your donation directly to us for another large group donation in honor of Jack or you could certainly send a donation directly to CRN.

Allowing us to tell our story of Jack, cystinosis, and the Cystinosis Research Network hopefully helps all of us to make a difference. In the spirit of St. Patrick's Day and in celebration of Jack's 5th Birthday, we thank you from the bottom of our hearts for everything and wish you and your families nothing but the best.

Sincerely,

Dave "Jack's Dad" Greeley

Christy "Jack's Mom" Greeley



Jack proudly smiles after pumpkin carving.  
(October 2004)



Jack and Alex enjoy winter at their grandparents' house.  
(December 2004)