

CYSTINOSIS  
RESEARCH NETWORK

*Dedicated to a Cure.*

*Committed to Our Community.*

# Cystinosis Parent Handbook



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# Using This Handbook

This handbook is a tool to use in your journey after a diagnosis with cystinosis. You are likely filled with questions about the disease and your child's future. This handbook will help you get a better understanding of cystinosis and its treatment, learn to care for your child, and find support with others in the cystinosis community. You may want to keep this handbook as a reference over the coming years.

Though getting a diagnosis of cystinosis is difficult for a parent, know that things will be OK. You have every reason to feel positive about your child's future. We will partner with you on your journey.

If you have questions, please contact the Cystinosis Research Network (CRN) at:

**Cystinosis Research Network**  
**302 Whytegate Court**  
**Lake Forest, IL 60045 USA**  
Web: <http://www.cystinosis.org>  
Phone: **847-735-0471**  
Toll Free: **866-276-3669**  
Fax: **847-235-2773**  
Email: [info@Cystinosis.org](mailto:info@Cystinosis.org)

The Cystinosis Parent Handbook was written with input from many members of the cystinosis community and the following contributors of the Parent Handbook Committee: Jean Blum, Maya Doyle LCSW, Christy Greeley, Colleen Hammond, Lauren Hartz, Jessica Britt Jondle, Marybeth Krummenacker, Katie Larimore, Mack Maxwell, Frankie McGinnis, Terri Schleuder, Steve Schleuder, Serena Scott, Sue Scott, Garrett Thomas, Lynn Thomas, Pam Woodward, Tahnie Woodward, and Jen Wyman.

The Cystinosis Research Network is an all-volunteer, non-profit organization dedicated to supporting and advocating research, providing family assistance and educating the public and medical communities about cystinosis. The Cystinosis Research Network's vision is the acceleration of the discovery of a cure, development of improved treatments and enhancement of quality of life for those with cystinosis. We are a private, nonprofit 501(c)(3) corporation, Federal Tax ID 04-3323789.

Edited September 2018

## MY CHILD WAS JUST DIAGNOSED WITH CYSTINOSIS.

### What Do I Do Now?

The initial period after a child is diagnosed with cystinosis is a tumultuous time. You may have experienced a long or traumatic "diagnostic journey" to discover what is making your child ill.

Learning that your child has a lifelong chronic disease feels life shattering. It is certainly life changing. But you can now begin to equip yourself and your family with the tools you will need to manage your child's disease and begin living with cystinosis.<sup>[1]</sup>

### Experienced Families Describe these Three Things as Vital:

**Knowledge:** Ask, watch, study, and read everything about cystinosis you can get your hands on. You are going to have many questions, and there is a lot to learn. Don't rely on memory, because it may fail you in times of stress. Create a binder with information about your child, cystinosis, medications, and the medical team. Write down questions in a pocket-sized notebook or on your computer as they pop into your head. Keep the notebook or computer with you and pull it out each time you see a doctor.

**Support:** Get involved with support groups. Connect with other families in the cystinosis community. Nobody knows a road better than somebody who has already been down it. Surround yourself with people who can guide you as you forge your own path.

**Planning:** Getting through daily life with cystinosis can be overwhelming. Become familiar with all of the things that will be needed to treat cystinosis. Make a written plan about how you will include activities such as medications and therapy treatments in your daily routine. This guide contains many suggestions for organizing your child's medical information and getting through everyday events in your child's life.

Sometimes life's most difficult challenges also provide its greatest gifts. In the beginning, cystinosis seems overwhelming, but with time, patience, and support, it can be managed. The planned 'journey' may have changed a bit, but a quality-filled life is possible.



## WELCOME TO HOLLAND

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*I am often asked to describe the experience of raising a child with a disability – to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this.....*

*When you're going to have a baby, it's like planning a fabulous vacation trip – to Italy. You buy a bunch of guidebooks and make your wonderful plans. The Coliseum. The Michelangelo David. The gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.*

*After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands. The stewardess comes in and says, "Welcome to Holland."*

*"Holland?" you say. "What do you mean Holland? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy."*

*But there's been a change in the flight plan. They've landed in Holland and there you must stay.*

*The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place.*

*So you must go out and buy new guide books. And you must learn a whole new language. And you will meet a whole new group of people you would never have met.*

*It's just a different place. It's slower-paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around.... and you begin to notice that Holland has windmills, and Holland has tulips. Holland even has Rembrandts.*

*But everyone you know is busy coming and going from Italy... and they're all bragging about what a wonderful time they had there.*

*And for the rest of your life, you will say "Yes, that's where I was supposed to go. That's what I had planned."*

*And the pain of that will never, ever, ever, ever go away... because the loss of that dream is a very, very significant loss.*

*But... if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things ... about Holland.<sup>[2]</sup>*

## EMOTIONALLY SURVIVING THE DIAGNOSIS

The news that your child has cystinosis can be heartbreaking. You may feel like it is impossible to escape feelings of anguish and loss in those early weeks and months while you grieve the loss of your dream of a healthy child. You may feel a range of emotions and reactions, including anger, sadness, and guilt. Whatever your feelings, they are normal, and you are not alone.<sup>[3]</sup>

Many parents have been in your shoes and are now travelling this journey with you. You will most likely travel this early diagnosis phase to a place of hope and happiness, a place where your family builds memories of love, laughter, and togetherness – despite cystinosis. Most parents have found help by creating a support group of family, friends, cystinosis support groups, spiritual leaders, and professional counselors.

### Be on the Lookout

Following diagnosis, parents can expect an emotional response similar to bereavement – shock, denial, anger, adaptation, and acceptance. Should any of the following responses embed themselves in your daily life and stay, however, you may want to consult with your primary care physician for a referral to a counselor who can help you work through what may very well be depression:

- Loss of interest in normal daily activities
- A sense of sadness
- Guilt
- Difficulty concentrating
- Difficulty sleeping
- Crying spells for no apparent reason
- Problems making decisions
- Irritability

### Coping and Hoping

As you develop a strategy for coping with the diagnosis of cystinosis, you'll need to begin planning for the immediate and long-term future. Children with cystinosis can plan for life as adults, but not because the disease has changed. The reason that people with cystinosis are living longer, more productive lives is because we've learned better ways to treat cystinosis. Much research continues to be done to improve treatment and find a cure, so it is important to stay hopeful. At the same time, preventing or slowing damage to the body, with consistent treatment from an early age, is the only way to give children with cystinosis a chance to grow and become adults. This is a big responsibility for you as parents and involves some sacrifice, but managing your child's regimen and teaching them to care for themselves over time promises the best outcomes.



## Some things you can do to ensure that your child has the best future possible:

**Establish a Routine:** Treatment of cystinosis requires taking medications frequently throughout each day. Fitting these things into your life can be a challenge, but consistency is the key to success. Having a scheduled time for everything can help ease the burden.

**Learn to Juggle:** Many parents of children with cystinosis find that they become so consumed with their child's needs that they forget to take care of their own. Don't let this happen, or you will quickly run out of steam. Cystinosis is a lifelong commitment. You'll need to find ways to make cystinosis a part of your life, without letting it be your life. You'll need to juggle your child's needs with your own, and with the rest of the family's.

## Settling in: Discovering a New Kind of Normal

It's hard for anyone to define what a "normal" life is, but clearly yours will have changed. Many families described coming to a "new normal" as they get accustomed to the management of cystinosis. There will also be surprises, and sometimes crises along the way. Your child, diagnosed when very young, may see taking medicine and other care tasks as normal. Teens and adults with cystinosis say "it's just what I've always done." With a few tweaks here and there, the definition of "normal" for people with cystinosis can be very similar to the definition of "normal" in a healthy person.<sup>[3]</sup>

### People with cystinosis can:

- Go to school, have friends, and play sports
- Get married, have a family, and have a career

When figuring out what normal means for you, the rule of thumb should be to dream big but proceed with caution. Set short-term goals to help you accomplish your life plan, but be flexible. You may need to make some adjustments along the way.

## Caring for Siblings



When a child is diagnosed with cystinosis, siblings need parents to honestly explain how cystinosis affects their brother or sister. Siblings need to know cystinosis is not contagious. Learning to live with cystinosis can be stressful for everybody involved,

but it is often terrifying for siblings who may be too young to understand what is causing the turmoil in their lives. Even older siblings sometimes have feelings of fear that their brother or sister will die. The unaffected siblings of a child with cystinosis may feel guilt for being healthy or for resenting all the time and attention the child with cystinosis gets from mom and dad. This can lead to acting out behavior or depression. Parents need to recognize signs of serious sibling reactions and seek professional help if it is beyond their ability to handle it.<sup>[5]</sup>

Although taking care of your child with cystinosis can be time consuming, it is important to spend "alone" time with each of your children.

### A resource for parenting siblings of chronically ill children is:

Siblings of Children Who Have Chronic Illness or Disability: Pointers for Parents by Cate Walsh Vockley, MS, CGC [http://www.nnpdf.org/familyservices\\_23.html](http://www.nnpdf.org/familyservices_23.html). Although this was written for Niemann Pick disease, it also applies to cystinosis.

Additional sibling resources are available on the CRN website.

### Build a Support Network

You and your family are not alone. Family support and community building is essential to individual and family long-term health and success. Contact CRN to connect with families who have walked in your shoes. Plan to attend regional family gatherings and family conferences.

CRN hosts family/physician conferences every two years to facilitate opportunities for parents, as well as children, teens, and adults with cystinosis to communicate and bond with one another.

Conferences and support groups provide opportunities for community, education, advocacy, and empowerment. Extended families members are always welcome. Eventually, you will know families from around the country and the world who are also living with cystinosis.

There are places online to discuss the various aspects of cystinosis and how it affects our lives, share tips about how we cope with it, vent our frustrations, and share our fears, our hopes, and our dreams.

Examples of support groups for parents, affected adults, caregivers, family, and friends:

#### Cystinosis Support Group

<http://www.cystinosis.org/support-groups>

#### CRN Facebook Group

<http://www.facebook.com/groups/6382741905/>

Researchers and medical professionals are very active participants in CRN, so you can reach out to experts and also encourage new healthcare providers to reach out for information.

### How Do I Talk to Others about Cystinosis?

Explaining cystinosis or medical procedures to a child can be hard for a parent who may still be coping with the magnitude of the diagnosis itself and dealing with their own feelings and fears.

Parents need to educate themselves about this disease so they can better manage symptoms, follow the care regimen, make informed choices about medical care, and answer their child's questions about it. In general, simplicity and honesty are the best policies. As your child gets older, and

as they show you they are ready by the questions they ask, you can provide more complex answers.

Your healthcare team can help both you and your child understand cystinosis in a variety of ways. Pediatricians or specialist physicians, nurses, social workers, and psychologists will all have their own way of talking with your child. Child life specialists, who work in most children's hospitals and many specialty practices, are specially trained in medical and therapeutic play and other interventions to prepare children for medical procedures and help them understand how their bodies work, what to expect about medical procedures, and what the nurses and doctors are doing.

An excellent resource for kids 6-12 years old is the book entitled *Nephropathic Cystinosis Explained to Children*, produced by Orphan Europe and the Cystinosis Foundation. In a very simple way, it describes the complexities of this disease. To learn how to obtain a copy of this book, visit <http://cystinosisfoundation.org>.

Throughout this guide, we will provide examples, by age, of how families have explained cystinosis to their child. Below, families volunteered their stories of how they explained and involved their support network in their child's new diagnosis

#### Explaining Cystinosis: Sarah

*Sarah was only 15 months old at diagnosis and she really did not understand much, so I guess that part of having to tell her was not a problem. She has grown with the knowledge and it is just her "normal." As for the other kids and our extended family, we just came right out with it. At the time of diagnosis Sarah was very sick. We were devastated at the diagnosis but also so relieved that we could tell our family that we have a battle plan. I gave my relatives a card with CYSTINOSIS written on it because nobody had ever heard of it and told them all to Google it! We have had a very open approach to discussions ever since. Our family policy is to hide nothing from Sarah or the other kids. We have found that knowledge is empowering for all of us. We feel a bit more in control of an uncontrollable situation.*

### Explaining Cystinosis: Sam

*We educated our family by creating an online website/blog about our son so that our family members and close friends could see his progress. We posted a simple definition of cystinosis and then we added links to cystinosis websites and videos for those who wanted more information.*

*My husband has two older cousins with cystinosis so a lot of our family members were familiar with the disease already. However, since their experience was 30 years ago, we were sometimes expected to do exactly as their parents did. So we gathered some members together to talk about concerns, like G-tubes, Carnitor, eating issues, Cystagon®, etc. It took a while for some family members to accept Sam getting a G-tube. I think the most helpful thing to do is refer the struggling family members to other blogs/websites where they can read about other children's experiences.*

### Explaining Cystinosis: Landon

*Landon is our only child. He went into the hospital with a plan to do an "intense feeding evaluation" because he wasn't eating and there seemed to be "no medical reason." Our family is local so they were there with us from day one as we learned bit by bit what was going on and that he may have cystinosis and what that meant. They learned with my husband and me.*

## FINDING INFORMATION AND EXPERTS

Information is power, and parents need to be strong advocates for their child with cystinosis. Read the articles on the CRN web page, and use a site like WebMD to look up words you don't know. Over time, the information will start to make sense and you will begin to understand more about this disease. Veteran parents will tell you, though: there is also more to learn as your child grows up!

Knowing all you can about cystinosis and your child's specific situation is beneficial when communicating with your child's doctors, specialists, and teachers. Think like a reporter: Ask questions like, "who, what, where, when, why, and how." Listen carefully to the answers you receive, and write down responses in a notebook. This will help if you need to explain this information to your partner or other involved caregiver.

### Build Your Medical Team

Start with the doctors you know first, perhaps those who diagnosed your child or saw you through a crisis. Do you trust them? Are they knowledgeable about cystinosis, or willing to learn and consult with experts? Do they have a multidisciplinary team to support you – such as a nurse practitioner, social worker, psychologist, nutritionist, and child life therapist? Working out of an academic medical center or children's hospital may give you access to more specialists and services, but being able to work with someone close to home in a smaller practice may also work for you.

Next, check for doctors in your local/regional area. Some families have a pediatrician who is very involved, while others coordinate their care through one of their specialists, particularly a nephrologist, geneticist, or metabolic specialist. One of these physicians should act as the team leader, understanding the big picture, ordering and running your cystine levels, and collecting all other doctor's reports. Having one doctor know what is going on with all the specialists is so helpful to all concerned. In the next section, we'll talk about making a care binder for your child, because you also need to have information at your fingertips. Ask your team to provide an emergency care letter to have available if unexpected visits are needed to emergency room or outside physicians. This will help maintain the integrity and consistency of your child's care, as will your vigilance!



If your insurance plan requires referrals, get them set up as soon as possible, and ask for the longest period of time allowed (a year at a time vs. a month at a time).

Your child will be working with these specialists for many years to come. You will be building a team over the years that may include other specialists such as ophthalmologists, gastroenterologists, endocrinologists, and nutritionists. There may be supplemental services, including physical, rehabilitation, occupational, speech, and eating therapy.

## CYSTINOSIS STANDARDS OF CARE

The newly developed Cystinosis Standards of Care and many excellent medical journal articles are available on the CRN website. Also on the CRN website is a list of medical professionals who have treated children, adolescents, and adults who have cystinosis.

### Participating in Research

Advances have been made in the understanding and treatment of cystinosis because many families and individuals have been willing to participate in research studies. People with cystinosis can play a critical role in the development of new treatments and the search for a cure by participating in research studies. To help promising drugs move swiftly from the research and testing phase to the people who need them most, many people with cystinosis are participating in clinical trials.

Research studies seek to answer questions about the understanding of cystinosis, new potential drugs, or new ways of using already approved therapies to treat cystinosis. Participating in a research study can be a very satisfying and worthwhile experience. All research studies that involve patients routinely undergo vigorous safety assessments to minimize the risks to participants before a clinical trial can begin. All studies are carefully monitored so that they can be stopped or altered very quickly if unforeseen problems occur.

### Additional Resources

- Learn about children and clinical trials:  
<http://www.nhlbi.nih.gov/childrenandclinicalstudies/index.php>
- Visit the CRN Research Participation site:  
<http://www.cystinosis.org>
- Visit the Cure Cystinosis International Registry:  
<https://cystinosis.individualcrossroads.org/>
- Visit the NIH Clinical Trials website to view current and complete studies:  
[http://clinicaltrials.gov/ct2/results?cond="cystinosis"](http://clinicaltrials.gov/ct2/results?cond=)
- The cystinosis experts at the NIH are available to answer questions and provide support to your team. To contact the NIH Cystinosis program, Dr. William A. Gahl and Dr. Galina Nesterova, email [bgahl@helix.nih.gov](mailto:bgahl@helix.nih.gov). Joy Bryant, RN, can be contacted at: **301-443-8690** or email: [bryantjo@mail.cc.nih.gov](mailto:bryantjo@mail.cc.nih.gov).

## WHAT IS CYSTINOSIS?

It is helpful to learn and remember the wording of a definition that is comfortable for you. The basic medical definition of cystinosis is: Cystinosis is a rare disease that is usually diagnosed in childhood. Cystinosis is a genetic metabolic disease that causes an amino acid, cystine, to accumulate in various organs of the body. Cystine crystals accumulate in the kidneys, eyes, liver, muscles, pancreas, brain, and white blood cells.<sup>[6]</sup>

It is estimated that at least 2,000 individuals worldwide have Cystinosis, though exact numbers are difficult to obtain because the disease is often undiagnosed or misdiagnosed. Cystinosis is estimated to occur in 1 out of 100,000 to 200,000 live births.<sup>[6]</sup>

### What Cystinosis is Not

Cystinosis is NOT contagious. Cystinosis is a hereditary disease (passed on from your genes) that cannot be passed from person to person by any form of personal contact like coughing or touching. It can be passed genetically, should your child have a baby. All children with cystinosis can have friends, go to school, do homework, play sports, exercise, and have fun.

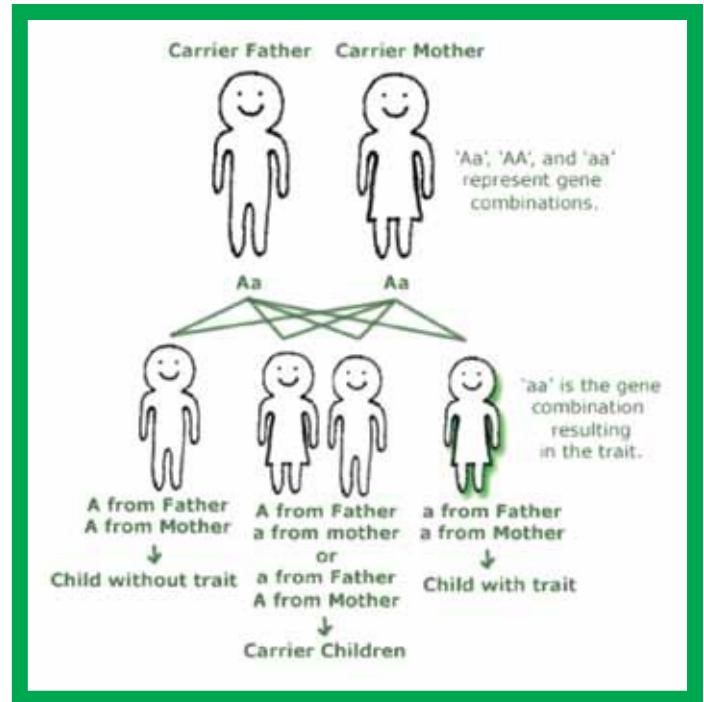
## HOW IS CYSTINOSIS INHERITED?

Every person's body is made up of millions of tiny structures called cells. Each cell comes with a full set of instructions, which tell the cell what to do and how to make our bodies work. The instructions are called genes, and they are made from a chemical called DNA. Genes usually come in pairs, and they determine everything about our bodies. For example, certain genes determine the color of our eyes, while other genes determine our blood type.

Genes are often called the “units of heredity” because the information they contain is passed from one generation to the next. We all get one gene in each pair from our mother and the other gene in the pair from our father. In this way our bodies work with a combination of instructions inherited from both our parents. Parents have no control over which genes get passed to their children.

Cystinosis is called a recessive genetic disease because parents do not exhibit symptoms, but they each carry a recessive gene, which may cause cystinosis in their children. The genetic path of cystinosis is therefore impossible to predict, and a cystinotic child is almost always a shock to parents. The recessive gene may lie dormant for many generations until suddenly two people with the defective gene have children.

Each time two such cystinosis carriers have a child together, there is a 1-in-4 chance (25% risk) of having a child with cystinosis. Every healthy sibling of a child with cystinosis has a 2-in-3 chance (66% risk) of being carrier, like his parents.<sup>[7]</sup>



## TYPES OF CYSTINOSIS

### There are three forms of cystinosis.

Infantile nephropathic cystinosis is the most severe form of the disease. Children with cystinosis appear normal at birth, but by 10 months of age, they are clearly shorter than others their age. They urinate frequently, have excessive thirst, and often seem fussy. At 12 months, they haven't walked yet and hesitate to bear weight due to pain from rickets. One of the major complications of cystinosis is renal tubular Fanconi's syndrome, a failure of the kidneys to reabsorb nutrients and minerals.

Generally, children with infantile nephropathic cystinosis are picky eaters, crave salt, and grow very slowly. If left untreated, this form of the disease would lead to kidney failure by 10 years of age. Thankfully, being left untreated is becoming less and less likely in the US and developed countries. Worldwide, however, there remain challenges to early diagnosis and access to treatment. In people with

intermediate cystinosis or juvenile (adolescent) cystinosis, kidney and eye symptoms typically become apparent during the teenage years or early adulthood. In benign or adult cystinosis, cystine accumulates primarily in the cornea of the eyes.

### How is Cystinosis Diagnosed?

The diagnosis of cystinosis is confirmed by the measurement of elevated level of cystine in white blood cells (WBC). The determination of the WBC cystine level is the key measurement for the diagnosis of nephropathic cystinosis and for monitoring the effectiveness of the treatment.<sup>[6]</sup>

The prenatal diagnosis of cystinosis for a child determined to be at risk for cystinosis is confirmed by measuring the cystine level in chorionic villus sampling or amniocentesis.<sup>[6]</sup>

## LIVING WITH CYSTINOSIS: LANDON'S DIAGNOSIS



Landon was born on April 8, 2011 weighing 7lbs, 4oz and was thought to be perfectly healthy. He was never a big eater, but grew well for the first 6-7 months of his life. Then his pediatrician noticed that the growing slowed down and then stopped around 9 months. She coached us on how much formula he should be consuming. He seemed to enjoy solids for a while but then around 10 months, he seemed less interested in his food. We were referred to a gastroenterologist who could find nothing medically wrong so ordered several tests to check for a variety of GI-related issues as well as Cystic Fibrosis. We were referred to a nutritionist and feeding team and although they made wonderful recommendations, nothing worked. Then, Landon began vomiting. We were sure that he had acid reflux, and if he had silent reflux, not wanting to eat would make sense. The gastroenterologist ordered a scope right before Landon turned 14 months. Again, she found nothing. She decided to admit him to Children's Hospital of Pittsburgh for an intense feeding therapy on June 2, 2011. Her hope was that by having a team to work on feeding issues with him on an individual basis, that we might figure out why he was not eating and work on a solution.

When Landon had the scope done, the gastroenterologist decided to collect a urine sample. She ordered another urinalysis upon

admission to the hospital. As a result of this urinalysis, Landon was diagnosed with Fanconi's syndrome. The gastroenterologist explained that sometimes Fanconi's syndrome stands on its own, but oftentimes it is a result of a metabolic disorder. After reviewing all of the possibilities, a rare disease called cystinosis made the most sense. One week later, the blood test that was sent to a laboratory in California indicated that our baby did, in fact, have this rare disease called cystinosis. Landon was hospitalized for 25 days before we were sent home, our lives forever changed.

### How the Cystinosis Diagnosis Affected Us

Cystinosis absolutely rocked our world, but we were thankful for answers. At the beginning, it seemed as if we went through all of the stages of grief over and over again and sometimes all in the same day! We still go through some of those stages at times, but the bad days seem to be less and less. It has made us feel more vulnerable. If this can happen to our child and our family, anything can happen. On the other hand, we have found strength in ourselves and in each other. We are learning how not to let cystinosis consume our lives. We are involved in fundraising and we have connected with hundreds of people who are also affected by this disease, but Landon is much more than cystinosis. He is a normal little boy who has some special needs.



## LIVING WITH CYSTINOSIS: JACKSON'S DIAGNOSIS



Hi, I'm Jackson and I'm 2 ½ years old. I was born in September 2009, perfect in every way. My first 8 months of life were awesome: sleeping through the night by 11 weeks (you're welcome, Mom and Dad), maintaining 90th percentile in weight and height, and meeting and exceeding every milestone that Momma's books had for me. Then, it was time for big boy food. I loved baby food, so solids should be fun, right? Wrong... Ugh, I gagged on almost everything I tried. Then I started puking... a lot. Then I stopped growing and got skinny. The first doc said that I had a bad gag reflex (if only!), then milk allergies, and Eosinophilic Esophagitis (huh?). Of course nothing made me feel better, I was spending more time in doctors' offices than at play dates, and Mom and Dad were going nuts. Finally one day we went to this scary place called the ER. We ended up living there for 10 weeks. I spent my first birthday and Thanksgiving there and started getting fed through my belly, taking lots of medicine, and getting poked in my arm a lot (owl!). I puked SO much, but oh well, I got lots of attention and was still happy every day.

I've overheard Mom and Dad talk a lot about what they learned while we lived in the hospital. It sounds like most people didn't know what I needed to get better. Mom and Dad had to get very pushy with a lot of people to make me better, and they kicked a lot of doctors out if they didn't really know what they were doing with me. After many, many doctors, they finally found the best belly doc (GI), and the best kid-knee doc (Nephrologist), but I'm not sure why I need a doc for my kid-knees! They also talked a lot about having to figure stuff out on their own. It seems like everything is trial and error with me. How much can they feed me at once (bolus) without making me throw up? How slowly do they have to increase my medicine so that I can tolerate a bigger dose? If my doc says that I need 3 more "mils" of bicitra a day, they will increase it by 1 for a week or two, then another for a few weeks, then another. They learned that the hard way, but I'm glad they figured it out. Momma says "Thank God for our cystinosis family", because since she's been talking to them on the computer she feels like she has somewhere to turn with questions, or if she just needs to vent. She also thanks God big time for KK (Kaitlyn), my nanny who lives with us. When I first met KK, puke freaked her out. I've helped her with that though, and now I can puke right at the dinner table and she'll still finish eating. I really love KK.

As for me, I've thrown up in all of my friends' houses, in the library, the mall, Target, Gymboree, the pool at swim class, the car (that poor car), my stroller, and pretty much anywhere I've been! Mom and dad have these cool buckets in different colors all around the house – I've put them to good use, too. Sometimes it comes out my nose and it hurts and I cry, but most times I just grab a drink of water and carry on with what I'm doing. Sometimes I wake up from my nap and my legs hurt, but I relax, have some water, and watch Dora and before I know it we've avoided Swiper, and used the map to climb the highest mountain and I'm ok again. I think that this cystinosis thing is much harder on Mom and Dad than it is on me. I don't know what the big deal is. I do everything that my friends do, actually sometimes even more. I'm in Swim, Art, and Gymboree. I know my letters, colors, shapes, and most of my numbers (there are only 10 of them, right?). Twice a week I play with Miss Dana (physical therapy) and I jump in the ball pit, climb the rock wall, and ride the "zipper" line. Twice a week I have lunch with my friend Betsy (eating therapy) and have so much fun. Pretty much everything in life is funny, even "time out". That seems to bug Momma sometimes – haha! I only throw up about every other day now, and lately Mom and Dad are having me drink my milk (formula) rather than putting it in my belly. It's OK, but I'd rather have water.

## TREATMENT OF CYSTINOSIS

Cystinosis is treated symptomatically. Renal tubular dysfunction requires a high intake of fluids and electrolytes to prevent excessive loss of water from the body (dehydration). Sodium bicarbonate, sodium citrate, and potassium citrate may be administered to maintain the normal electrolyte balance. Phosphates and vitamin D are also required to correct the impaired uptake of phosphate into the kidneys and to prevent rickets. Carnitine may help to replace muscular carnitine deficiency.

Beginning in 1967 technology became advanced enough to allow researchers a clearer understanding of the basic defect that defines cystinosis. They learned that the cystine level in plasma is normal, but is quite elevated within the cell. Additional studies showed that cystinosis is caused by a defect in lysosomal membrane transport. This defect causes a build-up of cystine within a part of the cell called the lysosome.<sup>[6]</sup>

Further research led to the discovery that renal transplantation is very successful in increasing the quantity and quality of life of a person with cystinosis. Because the disease is intracellular, researchers learned the transplanted kidney does not become “cystinotic”. However, the greatest advance in cystinosis research came in the 1970s when scientists discovered cysteamine (B-mercaphoethylamine) successfully removes accumulated cystine from the cells. For the first time this offered the hope of a future for those afflicted with cystinosis.<sup>[6]</sup>

### Cystagon®

Years of successful clinical studies through the '70s and '80s proved oral cysteamine not only removed intracellular cystine, but also preserved renal function and improved physical growth in children with cystinosis prior to renal transplant. In post-transplant individuals cysteamine preserves the function of non-renal organs such as the liver, pancreas, and thyroid. Cysteamine therapy is the treatment of choice for cystinosis individuals both pre- and posttransplant.<sup>[6]</sup>

Cysteamine (Cystagon®) has been approved by the Food and Drug Administration (FDA) since 1994 for the standard treatment of cystinosis. It is a cystine-depleting agent, meaning that it lowers cystine levels within the cells.

Cysteamine has proven effective in delaying or preventing renal failure, and also improves growth of children with cystinosis. In view of the harmful effects of chronic cystine accumulation, and the indications of the effectiveness of cysteamine therapy in various tissues and organ systems, oral Cysteamine should be used by post-transplant cystinosis individuals.

To successfully manage the symptoms of cystinosis, Cystagon® needs to be taken every six hours, every day. It comes in two doses: 150mg capsules and 50mg capsules. For younger children the capsules can be opened and mixed in food or liquid for easier administration. The dose is determined by the results of a specific blood test called a white cell cystine level, drawn 3-4 times per year. The target white cell (leukocyte) cystine content is less than 1.0 nmol of half-cystine per milligram of protein.<sup>[6], [8]</sup>

To attain the therapeutic level of cysteamine requires a gradual process of increasing the dose.

*“ For Cystagon®, start at a daily dose of 10 mg of free base per kilogram of body weight per day, given in divided doses every 6 hours and increased weekly or biweekly by 10 mg per kilogram per day as the drug is tolerated, until a target dose of 60 to 90 mg per kilogram per day is reached; target leukocyte cystine content is less than 1.0 nmol of half-cystine per milligram of protein and < 1.9 nmol nmoles per milligram of protein/m2 for the granulocyte assay. The recommended starting dosage of ProcySbi® for cysteamine-naïve patients is 0.2 to 0.3 grams/m2 per day divided into two doses given every 12 hours. A titration period of 4 to 6 weeks starting at 1/6 to 1/4 of the maintenance dose helps reduce the risk of side effects. Both preparations of cysteamine bitartrate max dose should not exceed 1.95g/m2/day. ”*

Cysteamine is not perfect. It has a sulfur-like smell and taste that causes unpleasant breath and body odor in most individuals. In some individuals it causes an increase in gastric acid, leading to serious stomach irritation and the need for daily treatment with medications called proton pump inhibitors that block acid production by the stomach. Though Cystagon® (cysteamine) has proven an effective way to remove cystine from most cells in the body, its effectiveness in removing cystine from the brain is still uncertain.<sup>[6]</sup>

## Cystaran™

Cysteamine eye drops dissolve corneal cystine crystals and relieve photophobia. Oral cysteamine does not remove cystine crystals from the cornea, but cysteamine eye drops administered 8-12 times a day has proven effective in clearing the cornea of the crystals. Leadiant Biosciences obtained FDA approval of cysteamine eye drops in October 2012 as Cystaran™ through a specialty pharmacy. Other options for the delivery of cysteamine to the cornea are under investigation by researchers.

## Fanconi's Syndrome

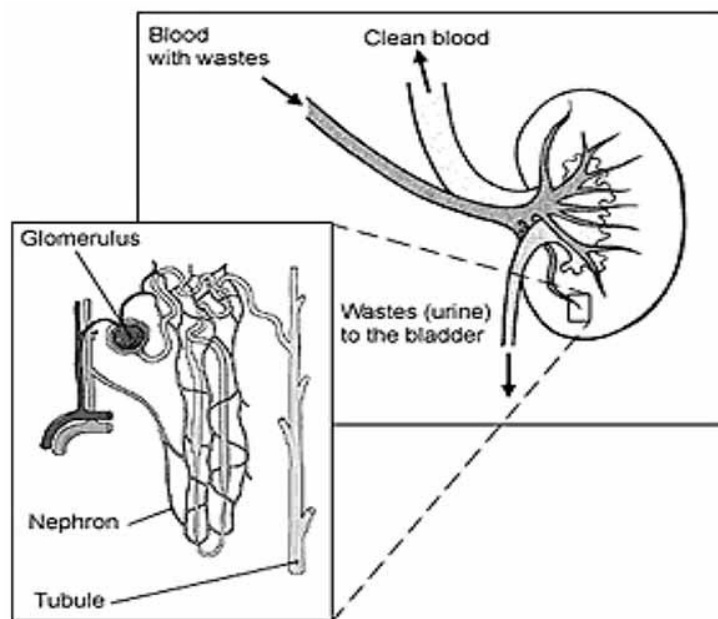
Cysteamine is the specific treatment of choice in cystinosis individuals, but it is not the only medication required. Supportive therapy is also needed. Cystinosis is the leading cause of Fanconi's syndrome in children. Fanconi's syndrome occurs when the kidney loses its ability to reabsorb water and various electrolytes necessary for normal body functions. Children with cystinosis have a narrowing in a portion of the kidney called the proximal tubule. This is referred to as a swan neck deformity and it is here that the kidney's ability to reabsorb water and various electrolytes is impaired. Children with cystinosis lose water, sodium, potassium, bicarbonate, calcium, chloride, amino acids, glucose, protein, and carnitine. Because of these losses, children with cystinosis need unlimited access to water. They usually crave salty and spicy foods such as pizza, pickles and potato chips. They require electrolyte replacement. Electrolyte replacement solutions may include sodium bicarbonate, sodium/potassium citrate, calcium supplements, sodium phosphate, and 1.25-dihydroxycholecalciferol.<sup>[6]</sup>

In the nephron (right), tiny blood vessels intertwine with urine-collecting tubes. Each kidney contains about 1 million nephrons.

In addition to removing wastes, your kidneys release three important hormones:

- Erythropoietin (eh-RITH-ro-POYeh-tin), or EPO, which stimulates the bones to make red blood cells.
- Renin (REE-nin), which regulates blood pressure.
- The active form of vitamin D, which helps maintain calcium for bones and for normal chemical balance in the body.

In Fanconi's syndrome the tubules do not completely reabsorb hormones, electrolytes, water, protein, etc. properly, and they are urinated out of the body.<sup>[6]</sup>





## Nutrition: Replacements, G-Tubes, J-Tubes And TPN

From the beginning, most children with cystinosis struggle with consuming enough calories to support growth and health. Their insatiable thirst for water due to the kidney defect described above fills their stomachs, decreasing the appetite for food. Frequent vomiting is also a problem for many children with cystinosis. When the diagnosis is made these children must also take numerous unpleasant tasting medications several times a day to manage symptoms. It can all quickly become overwhelming for the child and the parent/caregiver.

In some situations the doctor may decide another method of nutritional administration is best to ensure adequate caloric and medication intake. At this time the placement of a G-tube (gastrostomy tube) or J-tube (jejunostomy tube) may be discussed. Both a G-tube and J-tube are enteral methods of nutrition that involve placing a plastic tube through an opening in the gastrointestinal tract for the purpose of tube feedings. A G-tube goes through an opening in the abdominal wall directly into the stomach. If vomiting or reflux is severe or there is a problem with gastric emptying, a J-tube may be considered. A J-tube bypasses the stomach and is inserted through a hole made into a portion of the small intestine called the jejunum. Since there is no storage area in the jejunum, feedings must be given very slowly in small amounts. In both cases a special pump can be used to regulate the amount and rate of the feeding. The use of a G-tube or J-tube in children with cystinosis can be a short-term or long-term solution to feeding issues, but in most cases it should not be permanent. These enteral methods of nutrition should supplement oral nutrition not replace it. When the child becomes stable enough usually he/she is able to maintain health and growth through oral intake.<sup>[11]</sup>

There are many enteral nutritional supplements available. The child's physician will decide what best meets his/her needs. Supplements commonly used include Pediasure, Nutren, and Suplena.

In rare instances if the gastrointestinal tract is not absorbing nutrients for various reasons, a parenteral (Intravenous) method of providing nutritional and medication support may be considered. This method total bypasses the GI-tract and delivers nutrition through the circulatory system via a central

line catheter that is threaded through a vein ending at a vein close to the heart. This method of nutritional support is called hyperalimentation, Total Parental Nutrition, or TPN. This method is expensive, has a high risk for serious life-threatening systemic infections, and should only be used as a last resort.<sup>[11]</sup>

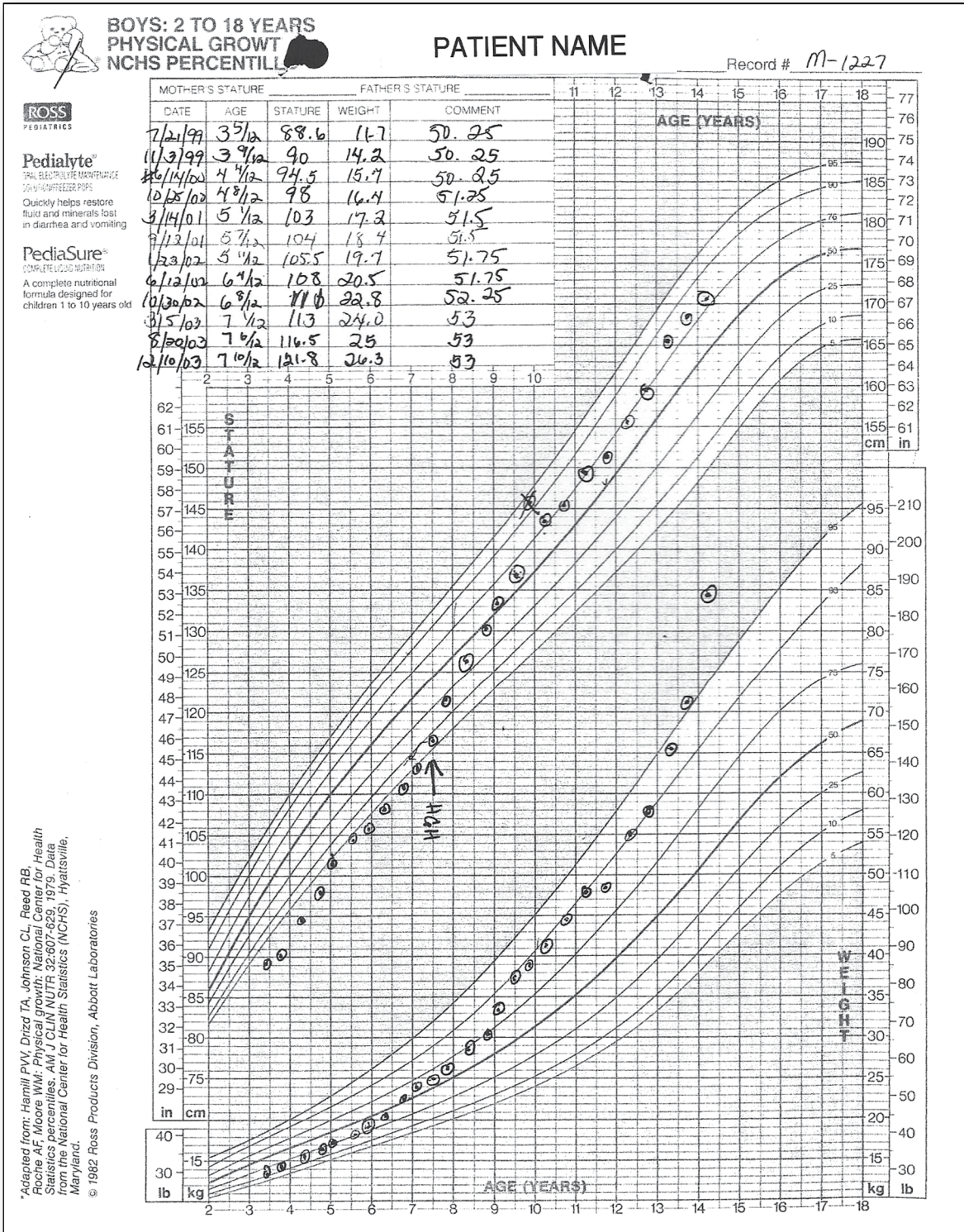
## Growth Hormone

One common and severe consequence many children with cystinosis experience is extremely slow linear growth. This occurs in part because of kidney disease and poor nutrition. Loss of Linear growth occurs, as does the Fanconi syndrome, in almost all patients with infantile cystinosis. The Fanconi's syndrome swan neck deformity removes the naturally occurring growth hormone during the blood filtering stage, and it is not reabsorbed.<sup>[12]</sup>

Children diagnosed with cystinosis and started on cysteamine before 24 months of age who are up to and above the 3rd percentile for height may benefit from the use of recombinant human growth hormone (rhGH) to improve linear growth. The combination of adequate cystine depletion with cysteamine, adequate nutrition and sufficient phosphate replacement are requirements for a normal growth rate, but seldom allow for catch up growth. Growth hormone is most effective before kidney disease advances to end stage renal disease (ESRD) and the need for dialysis or transplant arises. Some children, post-transplant, experience catch-up growth particularly on an immunosuppressive regimen that does not involve the use of steroids.<sup>[12]</sup>













There have not been sufficient studies to date to determine the effectiveness of growth hormone on individuals with cystinosis who have been transplanted and reached their maximum growth potential. Many in this group of individuals are now beginning to suffer from muscle wasting and muscle weakness, including difficulty swallowing. There is hope that future studies involving human growth hormone in this population may decrease the late symptoms some cystinotic teens and adults experience.<sup>[12]</sup>

GROWTH CHART OF MALE WITH CYSTINOSIS WHO STARTED GROWTH HORMONE AT AGE 7½ AND STOPPED WHEN X-RAYS INDICATED THE GROWTH PLATES WERE CLOSED





























## Common Medications for People with Cystinosis

Try [www.mymedschedule.com](http://www.mymedschedule.com) to keep track of medications, doses, times, and purpose for each medication.

MEDICATION	APPEARANCE	PURPOSE
Carnitor® (Levocarnitine)	 available in liquid or tablet	Treats carnitine deficiency
Cellcept® (Mycophenolate mofetil)		Prevention rejection
Coenzyme Q10		Supports normal heart function
Cozaar® (Losartan)		Controls blood pressure (angiotensin receptor blocker (ARB))
Cystagon® (Cysteamine Bitartrate)		Depletes cystine in the body
Cystaran™ Eye Drops (cysteamine ophthalmic solution 0.44%)		Removes cystine from corneas
DHT® (dihydrotachysterol)		Vitamin D. Also helps to absorb and use Calcium.
Diovan® (Valsartan)		Controls blood pressure (angiotensin receptor blocker (ARB))
Diamox (acetazolamide)		Used in treatment of Pseudotumor Cerebri
Dyazide (hydrochlorothiazide and triamterene)		Keeps Potassium from getting too low. Controls blood Pressure. Treats edema
Enalapril		Controls blood pressure; slows the process that leads to kidney damage (ACE inhibitor)
Epogen® (Epoetin Alfa)		Treats anemia



MEDICATION	APPEARANCE	PURPOSE
Humatrope® (Somatropin Recombinant)		Growth Hormone – Used in the body for the growth of bones and muscles.
Imuran® (Azathioprine)		Prevention rejection
Indomethacin		May help to decrease urine output
K-Phos Neutral (potassium phosphate and sodium phosphate)		Electrolyte replacement – phosphorus.
Kaon-Cl (potassium chloride)		Electrolyte replacement – potassium
Lisinopril		Controls blood pressure
Magnesium		Restores low magnesium levels
Multivitamin		Nutritional supplement
Myfortic® (Mycophenolate sodium)		Prevents rejection
Nexium® (Esomeprazole)		Treats/prevents stomach ulcer/heartburn
Norvasc (Amlodipine)		Widens blood vessels and improves blood flow. Treats high blood pressure.
Omega 3		Promotes cardiac and joint health
Pepcid® (Famotidine)		Treats/prevents stomach ulcer/heartburn
Potassium Chloride		Electrolyte supplement

MEDICATION	APPEARANCE	PURPOSE
Prednisone		Prevents organ rejection
Prilosec® (Omeprazole)		Treats/prevents stomach ulcer/heartburn
Procysbi® (cysteamine bitartrate) delayed-release capsules		Depletes cystine in the body
Prograf® (Tacrolimus)		Prevents organ rejection
Protonix® (Pantoprazole Sodium))		Treats/prevents stomach ulcer/heartburn
Reglan (Metoclopramide)		Speeds up stomach emptying
Rocaltrol® (Calcitriol)		Treats calcium loss from the bone
Sodium Bicarbonate		Antacid that neutralized stomach acid
Synthroid® (Levothyroxine sodium)		Replaces thyroid hormone
Urocit-K (Potassium Citrate)		Electrolyte supplement
Vitamin B-Complex		Treats Vitamin B deficiency/may help with Cystagon® odor Multi-Vitamin
Zofran (Ondansetron)		Used to combat nausea and vomiting

## Over the Counter Medications: What to Avoid and What to Use

- Tell your child's doctor about any herbs, supplements, or over-the-counter drugs your child takes. Just because some products are sold without a doctor's prescription does not mean they are safe for people with less than normal kidney function.
- Limit use of over-the-counter or prescription painkillers that contain ibuprofen (Advil®, Motrin®) or naproxen (Aleve®). These non-steroidal anti-inflammatory drugs (NSAIDS) cause blood vessels in the kidneys to shrink, so less blood flow comes through. If your child takes any of these drugs often, be sure to tell your child's doctor.
- Ask about a drug's effects on the kidneys any time your child takes a new medication. Some antibiotics are known to be hard on the kidneys. If you know that your child's kidney function is less than normal, avoid these drugs if you can and see if your child's doctor can prescribe something else.
- Avoid X-ray dye tests or have your child's doctor take steps to protect your kidneys. Less toxic dye can be used (this costs more), the dye can be diluted, and it can be flushed out of your body with extra fluid. Some doctors prescribe a drug called Mucomyst® to help protect the kidneys from the dye. (<http://kidneytrust.org/who>)

MEDICINE	USE	SAFE	AVOID
Tylenol – children's, liquid, tables, adult	Pain, fever	Discuss the dose with your doctor	
Delsym – 12 hour cough medicine	Cough	Safe	
Motrin	Pain, fever		Avoid
Over the counter cough syrup – multiple brand names	Allergies, flu, cold, cough		Avoid
Multivitamins	Supplements as vitamins	Generally safe. Discuss the brand and dose with your doctor	
Iron supplements	Anemia	Safe. Discuss the dose with your doctor	
Vitamin D	Rickets	Safe. Discuss the dose with your doctor	
Laxatives, multiple brands	Constipation	Use with caution. Discuss with your doctor	
Maalox, Tums, and others	Heart burns, Gastric reflux	Discuss with your doctor, i.e Zantac	Discuss with your doctor. Avoid Tums and Pepto-Bismol on dialysis
Fish oil	Some Improvement of bone and brain function, helps with high cholesterol	Safe	
Topical antibiotics (cream)	Skin infection or minor wounds or burns	Generally safe	
Eye drops (artificial tears or moisturizers)	Eye dryness	Generally safe	
Zinc Oxide lotion or cream	Diaper rash	Safe	

## Exercise

Though it is always good to check with a physician before beginning any exercise routine, a lifestyle that includes frequent cardiovascular and resistance exercise is a healthy choice for everyone. Children with cystinosis should participate in physical exercise such as gym class, outside play activities and recreational sports. Developing a lifelong habit of an active lifestyle will provide benefits of increased stamina, muscle strength, pulmonary function and cardiovascular health. Children with cystinosis do not sweat, salivate, or tear normally<sup>[1]</sup> so understanding your child's ability to cool themselves during exercise is important. Artificially sweating them, using a spray fan, or pouring water on their head and neck during hot weather, and intense exercise may be important for your child.

Post transplant there may be some activities to restrict. Specifically direct contact sports that could cause trauma to the transplanted kidney located in the lower abdomen i.e. wrestling and football. Most adults with cystinosis will experience varying degrees of muscle weakness, muscle wasting, and reduced pulmonary function at some point. While cysteamine delays the onset and perhaps the severity of these late symptoms, there is growing evidence in the emerging adult population who have been medication compliant that it does not eliminate them. Regular exercise in this adult population of cystinosis individuals may help maximize their physical potential.

An excellent example of an exercise regimen followed by one adult male with cystinosis can be reviewed on: <http://www.Cystinosisfoundation.org/macksworkout/macksworkout.html>.<sup>[13]</sup>

## Monitoring the Kidneys

For children with cystinosis, electrolyte imbalance and chronic kidney disease are some of the earliest complications. Frequent visits with a pediatric nephrologist (kidney specialist) become part of the "new" normal for a child with cystinosis. Monitoring the progression of kidney disease through frequent (monthly or weekly) blood tests also becomes routine. There are many resources to assist in understanding the blood test results. A helpful resource is: <http://labtestsonline.org/understanding/>

The child's blood pressure will also be watched closely. The occurrence of hypertension can increase as chronic renal

failure (CRF) advances. As renal (kidney) failure progresses, anemia may also occur. This is the result of a decrease in the production of a kidney hormone called erythropoietin needed to produce red blood cells (RBCs.) Medications such as Epogen or Procrit are available to stimulate RBC production if the level of erythropoietin is low. In recent years this has reduced the need for frequent blood transfusions in people with chronic kidney failure.

Chronic kidney failure in individuals with cystinosis follows a steady progression of decline until end stage renal disease (ESRD) is reached. Compliant dosing with cysteamine (Cystagon®) may increase the periods of kidney function stability and delay the onset of ESRD in cystinosis requiring the need for dialysis or transplant.<sup>[1]</sup>

## THE TIME FOR TRANSPLANT AND DEALING WITH DIALYSIS

Your kidneys are fist-sized organs, shaped like beans. They are located below your ribs, toward the back. Your kidneys



act as filters to clean waste (which are left over from breaking down food and your body's other activities) from your blood. Then they mix the waste with a little water creating urine. The urine goes to your bladder, which you empty

when you urinate. In addition to removing wastes from your bloodstream, the kidneys also make and regulate hormones and other chemicals in your body. When the kidneys aren't working correctly, your body can develop fatigue, bone problems, sleep problems, and anemia.

Cystinosis affects everyone differently. The age at which the kidneys fail and a transplant is required varies from person to person. Transplanted kidneys can come from living donors or deceased organ donors. Dialysis may be needed if a donor kidney is not available, or if a kidney transplant fails. A dialysis machine serves as an artificial filtering system that mimics a person's kidneys by removing waste from the blood.

The nephrologist or transplant team will be able to guide the family through the kidney transplant process. A transplant coordinator, social worker, or child life therapist may be able to offer ways to educate and prepare both individual



and family for an upcoming transplant. It may be helpful to speak with another family who has been through the dialysis and/or transplant process.

Although a new kidney will not be affected by Fanconi's syndrome or cystine accumulation, transplantation is not a

cure for cystinosis. Cystine will continue to build up in other parts of the body. Cysteamine therapy must continue after transplant.

## Kidney Transplant and Dialysis Resources

- American Association of Kidney Individuals
- American Kidney Fund®
- American Society for Transplantation
- Kidney School™
- Kidney and Urology Foundation of America, Inc.
- National Kidney Foundation®
- NIDDK Information Clearinghouse
- United Network for Organ Sharing

<http://www.aakp.org>

<http://www.akfinc.org>

<http://www.a-s-t.org/>

<http://www.kidneyschool.org>

<http://www.kidneyurology.org>

<http://www.kidney.org>

<http://www.niddk.nih.gov/index.html>

<http://www.transplantliving.org/>

## SOME BASICS ABOUT DIALYSIS

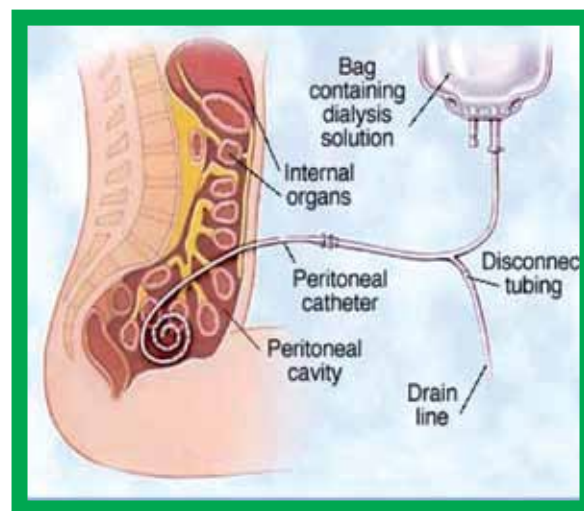
Dialysis is a therapy which eliminates the toxic waste from the body when the kidneys fail and cannot do their job. To date, all individuals with cystinosis will face ESRD (end stage renal disease) and the need for a kidney transplant. For some, an intermittent period on dialysis may be needed until a transplant is scheduled. The purpose of dialysis is to remove wastes, toxins and fluid from the blood after the kidney has lost its ability to function. Hemodialysis (HD) and peritoneal dialysis (PD) are the two types of dialysis.

### Peritoneal Dialysis

Peritoneal dialysis uses the body's own peritoneal membrane (abdominal lining) as a natural filter for blood. The dialysate fluid (cleansing solution) is infused into the abdomen through a catheter. Bodily wastes and excess fluid are passed into the dialysate fluid and after a time drained out. Fresh dialysate fluid is infused and the process begins again. Peritoneal dialysis allows more flexibility and privacy for the individual as it can be done at home, or wherever the individual is.<sup>[14]</sup>

There are three types of peritoneal dialysis. All use the same basic procedure, but the timing and process may vary.

- Continuous Ambulatory PD (CAPD) – The exchanges are done at home or wherever the individual is. The dialysate fluid remains in the body for 4-6 hours, is drained and immediately replaced.
- Continuous Cycling PD (CCPD) – A cycling machine does the exchanges over a 10-12 hour period each night while the individual sleeps. One or two daily exchanges may also be needed.
- Nighttime Intermittent PD (NIPD) – This type is similar to CCPD, but exchanges are made at night only, with no daily exchanges.<sup>[14]</sup>



## Hemodialysis

Hemodialysis (HD) is usually performed at a dialysis center 3 days a week, 3-4 hours a day. A machine is used with a special filter called an artificial kidney or dialyzer. After vascular access is established in the body through a port or an arteriovenous (AV) fistula, blood is passed through the dialyzer where it is cleansed filtered and returned to the body. The Nephrologist in charge will write a prescription for the amount of dialysis needed and monthly labs will determine its effectiveness. Prior to starting a hemodialysis treatment the individual is weighed, vital signs are taken and observations are recorded about the individual's physical status since the last treatment.<sup>[14]</sup>

In HD, blood is diverted from the body via the dialysis access (catheter or fistula) to a dialysis machine. The blood flows counter-current to a special solution called dialysate. The chemical imbalances and impurities of the blood are corrected and the blood is then returned to the body.

Some children undergo Hemodialysis in-center for 3-4 hours, 3 times per week while other families learn to provide hemodialysis at home.



## KIDNEY TRANSPLANTS

For many individuals with cystinosis, a kidney transplant is the treatment of choice once kidney disease has progressed to ESRD. During a kidney transplant a functioning kidney from a donor is surgically placed into a recipient whose own kidneys have failed. The transplanted kidney works to filter and remove waste products and regulate fluid levels in the recipient's body. Only one kidney is needed to replace the two diseased ones. The new kidney is placed in the lower abdomen and attached to blood vessels located there. The tube linking the new kidney to the bladder (ureter) is connected also. In most cases the native kidneys are not removed.<sup>[15]</sup>

Usually the improved quality of life after a successful kidney transplant is profound. In children, growth may increase dramatically, and energy levels are better. A kidney transplant recipient will require lifelong, daily immunosuppressive medication to prevent kidney rejection.

Improvements in these medications have reduced severe side effects significantly over the years.<sup>[15]</sup> Following a kidney transplant, the individual will receive many labs and be seen frequently for several weeks by the transplant team to monitor the functioning of the new kidney.

There are two types of donors for kidney transplants, living, either related or unrelated, and deceased. Family members are often the first choice for a kidney donor, but nonrelated donors also work if the blood and tissue types match closely enough. If a living donor is not an option the transplant candidate may be added to a waiting list for a cadaver kidney. This option may require a wait of one or more years.<sup>[15]</sup>

Transplant is NOT a cure for cystinosis, and children will need to continue Cysteamine and other needed meds, and begin taking a regimen of immunosuppressant drugs to maintain health and prevent rejection of the graft kidney.

## MUSCLE WEAKNESS

Prior to the widespread use of cysteamine as the treatment of choice for children with cystinosis, individuals survived into early adulthood solely because of kidney transplantation. In this group of individuals, the detrimental effects of years of cystine accumulation in various cells and organs became apparent. Researchers learned the accumulation of cystine crystals in the oropharyngeal (mouth and pharynx) muscles caused an impaired ability to swallow serious enough that many died from aspiration of food. Impaired speech was also noted in many. A weakening and wasting of skeletal muscle including in the hands was observed as a common late symptom.<sup>[16]</sup> For more information, the Cystinosis Standards of Care are available on the Cystinosis Research Network website at <http://www.cystinosis.org>.

With the discovery that cysteamine reduced cystine crystals in the kidney, thereby extending kidney function by many years, it was hoped other organs might also be spared with the use of cysteamine. Currently cysteamine is the treatment of choice for both pre and post-transplant individuals with cystinosis. Studies are on-going to determine if cysteamine therapy from early childhood will prevent the devastating late symptoms of cystinosis.

## PSEUDO TUMOR CEREBRI

Pseudo Tumor Cerebri (PTC) also referred to as idiopathic intracranial hypertension (IIH), has been diagnosed in a number of cystinosis individuals over the last 10-12 years. In the general population the incidence of PTC is 0.9 per 100,000 people. In the cystinosis population the incidence of PTC is 3-5 percent. All ages and both genders seem to be affected in the cystinosis population. PTC is caused by a build-up of cerebral spinal fluid in the cranium that puts increased pressure on the optic nerves. Unrelieved, this increased pressure can irreparably damage the optic nerves causing permanent vision loss including blindness. Frequent headaches are the primary early symptoms of PTC. Swelling of the optic nerves, called papilledema, observed on an

eye exam is followed up with a MRI of the brain. If the MRI is negative the next step in the diagnosis of PTC is a lumbar puncture to check the cerebral spinal fluid pressure (CSF). If the CSF pressure is elevated, the diagnosis of PTC is confirmed. There are medications available to treat PTC. Therapy includes an oral diuretic medication, which generally resolves the problem within months. Frequent eye exams by a neuro-ophthalmologist are necessary to monitor the visual acuity of the individual and the effectiveness of the medication in reducing the papilledema.<sup>[17]</sup>

There is speculation that because of the blood-brain barrier, cysteamine does not reach the part of the brain involved in the normal reabsorption of CSF. The presence of cystine crystals in this area may be a cause of the build-up of CSF leading to PTC in cystinosis individuals.<sup>[17]</sup>

# CREATING A CARE BINDER FOR YOUR CHILD

Many parents have binders to help them organize important information. A care binder has multiple uses. A binder helps parents maintain an ongoing record of their child's care, services, providers, and notes. The binder is a great tool in empowering families to become the experts on their child's care. It is also a way to maintain the lines of communication between the many providers and services that help care for a child and their family.

You can compile information for your child's binder collecting information on an ongoing basis.

- When your child has a diagnostic test or procedure, ask for a copy of the test results or procedure report.
- At each doctor appointment, ask for a copy of any new information added to your child's medical record. You may also want to write your own notes detailing what happened at the appointment.
- If your child spends time in a hospital, ask to take a copy of your child's medical record with you when you leave.
- Consider keeping a health diary to write down your child's symptoms and side effects of medications and treatments.
- Keep copies of medical bills and insurance claims.

## Monitoring Cystine Levels

Common to all cystinosis patients is the need for ongoing monitoring of mixed leukocyte cystine level X 3-4/year. This encourages adherence with cysteamine therapy and guides dose adjustment with changes in body size or in accordance with clinical goals. This special test requires establishing a local arrangement for timed, point-of-care isolation of mixed leukocytes from "trough" blood drawn in the morning just prior to the next cysteamine dose (6-8 hrs after the last dose of Cystagon or 12 hours after the last dose of Procysbi). The leukocyte pellet is then transferred to a reference laboratory for assay. The therapeutic target (about 15-20% of untreated baseline) is <1nmol "half-cystine"/mg protein.

Therapeutic response to cysteamine should be evaluated three to four times per year by the analysis of white blood cells. In the United States, cystine assays are performed at the University of California at San Diego (UCSD) Cystine Determination Laboratory and Baylor College of Medicine Medical Genetics Laboratory.

- UCSD Cystine Determination Laboratory and the Cystinosis Central website is a comprehensive resource for information about monitoring cystine levels: <http://www.cystinosiscentral.org/cystine.html>
- UCSD Cystine Determination Laboratory  
CTF - Bldg B Room 213  
212 Dickinson Street, San Diego, CA 92103  
Phone: 1-619-543-5260 Fax: 1-619-543-3565  
Email: [cystine@ucsd.edu](mailto:cystine@ucsd.edu)
- Baylor College of Medicine Medical Genetics Laboratories  
One Baylor Plaza, NAB 2015, Houston, TX 77030  
Phone: 1-800-411-GENE (4363) Fax: 713-798-2787  
[http://www.bcm.edu/geneticlabs/test\\_detail.cfm?testcode=4627](http://www.bcm.edu/geneticlabs/test_detail.cfm?testcode=4627)

**Reminder:** Blood must be taken 5-6 hours after a dose of Cystagon® and processed immediately for the white blood cell cystine measurements to be meaningful.



## Organizing and Storing Your Child's Care Binder

Many people keep at least part of their medical record on a computer. You can type notes on the computer and scan papers into a computer file. If you decide to keep your medical record online, you should print a copy to take with you to doctor appointments and regularly back up your record onto a CD or a removable drive.

However you choose to organize your personal medical record, keep it in a secure location, such as a safe deposit box or fireproof home safe. You may also want to give a copy of important information to a relative or friend in case of an emergency.

You may also maintain your personal medical record on the internet through a number of companies that charge a fee for their services. With your permission, records stored online can be accessed by family members and doctors.

Before choosing an online company, carefully check the security and confidentiality measures the company uses to protect your medical information.

## Creating Your Care Binder

The National Center for Medical Home Implementation has developed helpful templates to create a Care Binder for your child. Below is a list of downloadable forms with which you can build your own Care Notebook. You may download a complete section's documents by clicking on the section's ZIP link or select specific documents by clicking on the sub-category links below. All Care Notebook forms are made available in both Microsoft Word and Adobe PDF versions for your convenience.

The forms can be downloaded from:

[http://www.medicalhomeinfo.org/for\\_families/](http://www.medicalhomeinfo.org/for_families/)

### Forms available include:

#### Care Notebook Cover Page

##### Personal Information

- Care Providers
- Insurance Information
- Family Information
- Family Support Resources
- Funding Resources

##### Pages to Keep Track of Appointments and Care

- Appointment Log
- Diet Tracking Form
- Equipment-Supplies
- Growth Tracking Form
- Hospital Stay Tracking Form
- Immunizations
- Information Needed by Emergency Care Providers
- Lab Work-Tests-Procedures
- Make-a-Calendar
- Medical Bill Tracking Form
- Medical-Surgical Appointments
- Medications
- Family and Child Medical History

##### Personal Notes

- Parent and Child Questions for Doctor
- Parent and Child Questions for Setting up Home Care

##### Care Needs of My Child-Abilities and Special Needs

- Activities of Daily Living
- Care Schedule
- Child's Page - Now and Later
- Communication
- Coping-Stress Tolerance
- Mobility
- Nutrition
- Respiratory
- Rest-Sleep
- Social-Play
- Emergency Plan
- Baby-sitters Guide
- Information for Caregivers – Instructions for Care

##### Community Health Care-Service Providers

- Medical Dental
- Public Health
- Home Care
- Therapists
- Early Intervention Services
- Child Care
- Respite Care
- Pharmacy
- Special Transportation

##### School Issues

- School-Making it work
- Home-School Worksheet
- School Communication Sheet
- Permission for Procedures – Medications at School
- Physical Education Activity Guide
- Insert your Individual Education Plan or 504 Plan
- Transitions-Looking Ahead

# BINDER IDEAS FROM PARENTS OF CHILDREN WITH CYSTINOSIS

## Kacy's Binder

We have a 3-inch medical binder for Kacy. We have it divided into several sections: doctors and contacts, insurance, lab work and medical reports, medications and prescriptions, school, ophthalmology, articles and information, and transplant. We place everything that is medically related to Kacy in this binder. I try to update it before or after each visit to a doctor. Any tweaks made in medications or dosages, any new lab work, slit lamp eye exam photos, etc. get filed into place. I often think I can remember a date of a procedure or a medication change, but more often than not I am wrong and have to refer to our binder. Her doctors have borrowed it to read through. I think it might be more informational than her medical files in the office.

## Shea's Binder

Maintaining a binder with Shea's medical records was a great tool for me when Shea was a child and is now helpful for him as a young adult. We keep a "current" medical binder that is a one-inch binder. We keep many past medical history documents in three-inch binders. The "current" binder is an easy file to bring to doctors appointments and hospitalizations. We keep copies of the documents on our computer and amend as changes occur.

The binder tabs include:

- One-page medical form with copies to share with physicians as necessary. Please see sample shown.
- Immunization history
- Care providers page. We staple or scan physician/ medical professional's business cards to this page
- Pharmacy Information
- Insurance Information, referrals and pending bills
- Cystinosis Information
- Latest blood test results and blank lab request forms
- Notes page – Questions for medical appointments

One-Page Health Information Sheet			
BASIC INFORMATION	Name:		Physicians:
	Date of birth:		
	Address:		
	Home phone:		
	Cell phone:		Allergies:
			Immunizations:
	EMERGENCY CONTACT INFORMATION		INSURANCE INFORMATION
Name:		Primary:	
Phone:			
Relation:		Secondary:	
MEDICAL HISTORY	DATE	PROCEDURE / EVENT	REASON
	May 1983	Cystinosis diagnosis	
	August 1999	Kidney transplant	ESRD
MEDICATIONS	NAME	DOSE / TIMES	PURPOSE
	Cystagon	450 mg QID	Lowers cystine levels
	Tacrolimus	2 mg AM / 1 mg PM	Prevents kidney rejection

## CYSTINOSIS HEALTH JOURNAL

Jean Blum, mother of Jackson, developed a Cystinosis Health Journal Work Book template to assist with tracking the many needs of your child with cystinosis in an easy to access format. You are encouraged to customize the workbook for your own needs. The complete template is on the CRN website at <http://www.cystinosis.org>.

<b>Handout</b>	One page document to print and bring to every doctor appointment. You may want to keep a copy in your purse, briefcase, diaper bag, and glove box. Every time it is updated, email a new copy to all caretakers and replace the outdated printed copies information.
<b>Contacts</b>	Health care team information. You may want to print a copy and put on your refrigerator.
<b>Checklist</b>	Daily medication schedule and important reminders. This is handy especially if more than one person is administering medications throughout the day. Print multiple copies and keep on the refrigerator or counter. Put a check in the box once the medication is given and you don't have to wonder if it was done. You may want to put the exact times rather than morning/afternoon/evening.
<b>AM Checklist</b>	This handy checklist is not intended for every day use, but rather for the occasional sitter or caretaker. Customize it for your routine, print and cut out the four lists and they will be thankful that you prepared this for them.
<b>Labs</b>	This is good to see trends with the labs. Highlight any that were out of range.
<b>Timeline</b>	Track important changes, procedures, weight and height here.
<b>Symptom Tracker</b>	You may need to track vomiting or any other symptoms on a short-term basis to monitor changes with medications, formula, etc.

## PARENTING A CHILD WITH CYSTINOSIS

### Your Child's Development Across the Years

We will use the ideas of development psychologist Erik Erikson, and cognitive and educational expert Piaget to describe some of the basic stages of child development, and how these things might be affected by cystinosis. There is a wealth of information out there about normal child development, which you can use as a “yardstick” as your child grows. We will also include some information about specific cognitive and learning issues that have been noted in children with cystinosis, though your child may not experience these problems.

### Erikson's Stages of Development

Erikson described eight stages of development through which a healthy developing human should pass from infancy to late adulthood. In each stage the person confronts, and hopefully masters, new conflicts and challenges. These stages overlap and may not be the same for every child, but Erikson's model is helpful in understanding how your child is behaving and coping. <sup>[20]</sup>

From birth to age six a child typically works through the first three of Erikson's stages. They are: <sup>[20]</sup>

- 1. Trust vs. Mistrust, birth to 18 months (birth to walking)**
  - Children develop a sense of trust when caregivers provide reliability, care, and affection. A lack of this will lead to mistrust.
- 2. Autonomy vs. Shame and Doubt, 1 to 3 years (toddler, toilet training)**
  - Children need to develop a sense of personal control over physical skills and a sense of independence. Success leads to feelings of autonomy, failure results in feelings of shame and doubt.
- 3. Initiative vs. Guilt, 3 to 6 years old (preschool, nursery, exploring)**
  - Children need to begin asserting control and power over the environment. Success in this stage leads to a sense of purpose. Children who try to exert too much power experience disapproval, resulting in a sense of guilt.

Erikson's additional stages that relate to childhood are:

- 4. Industry vs. Inferiority, 6 to 11 years (school age)**
  - Children need to cope with new social and academic demands. Success leads to a sense of competence, while failure results in feelings of inferiority.
- 5. Identity vs. Role Confusion, 12 to 18 years (social relationships)**
  - Teens need to develop a sense of self and personal identity. Success leads to an ability to stay true to yourself, while failure leads to role confusion and a weak sense of self. <sup>[20]</sup>

## PIAGET AND CHILD DEVELOPMENT

Psychologist Jean Piaget's cognitive-developmental stage theory described how children's ways of thinking developed as they interacted with the world around them. Piaget's theory has four stages: sensorimotor, preoperational, concrete operational, and formal operational. <sup>[21]</sup>

- 1. Sensorimotor stage, birth to 2 years (beginning learners)**
  - Children figure out how to make use of their bodies. They do everything with their five senses, and by learning to crawl and then walk, point and then grasp.
- 2. Preoperational stage, 2 to 7 years (language)**
  - Children use mental symbols to understand and to interact with the world, and they begin to learn language and to engage in pretend play.
- 3. Concrete operational stage, 7 to 11 years (logical problem solving)**
  - Children begin to organize information they learn. They are limited to considering only concrete information. At this stage the capability for abstract thought isn't well developed.
- 4. Formal operational stage, 11 to adulthood (abstract thinking and problem solving)**
  - Adolescents learn how to solve abstract problems and to think symbolically. <sup>[21]</sup>

Parenting any child through these stages toward life as a healthy adult is a challenge, but even more so for the child with a chronic illness such as cystinosis. Maintaining family expectations, routines, rules, respect for others, encouraging self-reliance, and trying very hard not to over-indulge or over-protect the child is important.



It is very important to provide a normal life, granted, that “normal” life will be redefined through the lens of cystinosis. The new “normal” includes medicines, treatments, doctors’ visits, labs, and hospitalizations, but it should also include family activities, friends, community, faith, school, sports and recreational activities, chores, and age appropriate responsibilities. It is through all of these activities that one develops self-confidence, self-esteem, and the ability to relate to others - all needed to successfully navigate life with or without illness.

## INFANCY AND TODDLERHOOD

Erikson defined the stage from birth through age two years as “trust versus mistrust”. This stage centers on the infant’s basic needs being met by his or her parents, and this interaction leading to trust or mistrust.<sup>[20]</sup> The infant depends on the parents for sustenance and comfort. The child’s early understanding of how the world works and how people behave, develops from their interaction with their parents and caregivers. If the parents provide the child with warmth, regularity, and dependable affection, the infant’s view of the world will be one of trust (even in situations where other resources are limited). If parents and caregivers are consistent sources of food, comfort, and affection, an infant learns trust - that others are dependable and reliable.

For a child who is hospitalized or ill, and for whom things are unpredictable, this can be a challenging time. They will not understand what is happening to them rationally, but may react to the anxiety or stress they sense from parents. Consistency and calm may be the last things you feel capable of! Your “reliable” behavior, and building a regular routine filled with familiar things (whether at home or hospital) can help your child develop a sense of trust and safety. That trust will eventually extend beyond you to others in their lives – family, friends, teachers, doctors and nurses, etc. While the memories of your very young child being very ill, or that moment of diagnosis, may be blasted into your memory, your child will remember little, or just a little, of these traumatic experiences.

### Early Diagnosis and Treatment

Children with cystinosis appear normal at birth, but by 10 months of age, they are clearly shorter than others their age. They urinate frequently, have excessive thirst, and

often seem fussy. At 12 months, they haven’t walked and bear weight only gingerly. Due to Fanconi’s syndrome, minerals are lost in the urine, which must be replaced. Renal tubular dysfunction requires a high intake of fluids and electrolytes to prevent excessive loss of water from the body (dehydration). Children with cystinosis usually take electrolyte replacements around the clock. Generally, they are picky eaters, crave salt, and grow very slowly. In the early stages of diagnosis, many children with cystinosis vomit 5 – 10 times a day.

### “Something Is Wrong with My Child.”

Many parents share similar stories about how they knew something was wrong with their children and what led them to bring their children to the doctor. The child is on the normal growth curve until about age 6 months. The child starts vomiting multiple times a day. Parents are told their child has a virus. Vomiting continues and the child does not achieve developmental milestones of sitting up or walking. Their appetite decreases and the number of wet diapers increases. Parents have sleepless nights trying to comfort their child, cleaning vomit and changing diapers. Days are filled with many formula changes in a desperate attempt to get the child to eat. Parents return to the doctor again and again. Finally, the child is admitted to a hospital for dehydration or failure to thrive. A urine specimen reveals the child is spilling many electrolytes into their urine.

The child does not have diabetes. The child is diagnosed with Fanconi’s syndrome. A wrist x-ray shows the child has rickets. Doctors suspect cystinosis. Blood tests confirm the child has cystinosis. Parents are both devastated and relieved, that after months of worry, illness and testing, their child finally has a diagnosis.

If you have recently experienced diagnosis, you have learned a vital lesson: trust your instincts as a parent. Your knowledge about your own child and your observations will be key to managing this illness.

Parents keep journals and create binders as they navigate this new life. They divide shifts of giving medications. They track blood test results on excel spreadsheets. They seek out information and reach out to online support groups. After a while, they do adjust to their “new normal.” Life may never be the same, but families do learn how to live it – around, in spite of, and because of cystinosis.

## LIVING WITH CYSTINOSIS: LAURA'S DIAGNOSIS

Laura came roaring into this life on March 23, 1996, 8lbs. 15.5 oz. of screaming felinity. At birth, Laura was given 10 on APGARs and for all concerned was perfect. Perfect lasted for us a total of two weeks. This was when we had our first ER visit and our first specialist referral. Laura was having respiratory issues, spiking fevers of 103 and vomiting frequently.

Our first specialist was a gastrointestinal physician who suspected Laura may have cystic fibrosis. He placed her on two medications to help her food move quicker out of her stomach and to help with reflux. Laura was given every test known to man for eating and respiratory issues. Yes, she had delayed emptying of her stomach, yes she had asthma, yes she had reflux, and all were treated yet she continued to get sicker. That is the best term to describe her; she was a happy, beautiful little baby who just seemed to stand still in time size wise. Her development, other than teeth and standing, was completely normal, yet her size became more markedly delayed every day.

She was hospitalized at 11 months for a G-tube placement to try to get more nutrition to her, a simple procedure that we expected to take place in same day surgery. Our one-day ended up being a month. Laura did not react well to the withholding of fluids or to anesthesia. As her rollercoaster labs continued, someone mentioned a nephrologist should see her. Dr. Robson came into our room, looked at Laura, looked at her massive medical chart, and informed us he was ordering a blood test but he was 99% sure our daughter had cystinosis. We had no idea what this man was talking about, but after a quick search of the medical library and seeing the words "always fatal", and the horrible picture painted on the "world wide web" that existed in 1996, we knew he had to be wrong. He wasn't. Dr. Robson sat me down and handed me a notebook of every article ever published at that point on cystinosis. He informed me it was my duty as Laura's mother to be her advocate, learn as much as possible, fight harder than I ever imagined, and learn to get every available advantage for Laura, to see that she had the best life possible. He did not give me a moment for woe is me. He would not allow any pity, only an empowerment for which I will be eternally grateful.

## LIVING WITH CYSTINOSIS: LANDON'S STORY



Landon and our family have faced multiple challenges since he was diagnosed at 14 months. Learning and accepting our new "normal" is something that we continue to strive towards. Landon is so resilient and amazes us every day.

Landon is fed, at this point, mainly from a feeding tube. He gets bolus feeds throughout the day and a continuous feed for 10 hours at night. For the first six months, we struggled off and on with vomiting. We experimented with how much his belly can handle at once, whether we should slow the feed down or speed it up a bit. Another challenge we have faced is getting him to take in food orally. We have been consistent about offering him food at each meal, right before a feed, and we have initiated services with an early intervention group. An occupational therapist comes to our house once a week to work on feeding issues. For a few months, Landon showed very little interest in eating, but now he asks to eat all throughout the day. We did find that he holds food in the side of his mouth and doesn't swallow it. With time and some exercises, he is beginning to take in more and actually swallows. The feeding tube is not going away any time soon, but we are certainly making progress.

Adjusting his medication has also been a bit of a challenge. It is important to keep his electrolytes within a normal range, so he has to have blood taken regularly to see how everything looks. We have been fortunate in that Landon has tolerated his medication pretty well and his levels look pretty good for the most part, but there certainly has been some experimentation involved!

His gross motor skills were also a concern for us. Landon was on target for most skills except for walking. We also initiated early intervention services with a physical therapist. We are proud to say that our little guy is walking now and has made incredible progress in the past few months. We continue to work with the physical therapist to support him.

On a typical day, Landon usually wakes around 8 am. I wake one hour before to give him a medicine called Levothyroxine to keep his thyroid levels in a normal range. All of Landon's medications are given through his MicKey button. At 8 am, I start his first bolus feed of the day, which usually takes about one hour from start to finish. Around this time he also takes his first daytime dose of Cystagon® along with three other medications. Depending on the day, we may leave after this feed and go to our Mommy and Me pre-school program. We run errands, visit friends or grandparents, or just stay in the house and play.

Around 11:00 am, we have lunch. Some days he eats a little bit and other days he just plays with the food. At 12:00 pm, he goes down for a nap and starts his second bolus feed of the day and three more medications. He typically naps until around 2:00 pm and when he wakes, it's time for Cystagon® and one other medication. Two days a week he has either occupational therapy or physical therapy. At 3:30 pm, Landon is given something to eat and at 4:00 pm, we start his third bolus feed of the day and he takes four medications. The rest of the evening includes dinner for mommy and daddy while Landon sits in his high chair and participates in dinner with us. Then we have playtime, bath time, stories and bed at 7:00 pm. His feed starts at 7:00 pm along with given three medications. At 8:00 pm, he is given Cystagon® and one other medication. At 2:00 am, I wake to refill his formula and to give him Cystagon® along with one other medication. He stays asleep during this time.

During the feeds in which he is awake, we spend time watching the television shows that he likes, doing puzzles, playing with toys and reading books. His feeds and medication schedule never stop us from going anywhere or doing anything. It was a bit overwhelming at first, but now, it's just what we do.

## Developmental Milestones

Infants and toddlers with cystinosis typically experience illnesses related to electrolyte and gastrointestinal complications. Frequent illnesses and hospitalizations during this time may affect the child's ability to reach developmental milestones. Although each baby develops in her own individual way and at her own rate, failure to reach certain milestones may signal medical or developmental problems requiring special attention. If you notice (or just suspect) that your child is not reaching developmental milestones, contact your pediatrician. Your pediatrician will likely review development milestones at every visit. Information is available online at sites like [babycenter.com](http://babycenter.com) or WebMD, or in commercially available "baby books."

## Developmental Services Available to Families of Children with Cystinosis

Most states have early intervention programs to address the developmental needs of children from birth up to the third birthday; assessment and intervention is typically free or low cost. Your pediatrician should be able to refer you to early intervention services in your state or you can check with your state's Department of Health and refer yourself. These programs typically provide comprehensive, integrated services, utilizing a family-centered approach to facilitate the developmental progress of eligible children. Early Intervention services are designed to meet the developmental needs of each child and the needs of the family related to enhancing the child's development. Services may include speech, occupational and physical therapists, developmental educators, social workers, psychologists and nurses. In addition, early Intervention programs may contract with consultants in areas such as nutrition, adaptive equipment, and behavior management.

## EARLY CHILDHOOD, AGES 3 - 6

According to Piaget, children aged 3 - 6 are in the preoperational stage of cognitive development.<sup>[21]</sup> In this stage the child's memory and imagination are strong. They are very good at using symbols as evidenced in imaginative play. They may pretend to be mommy or daddy, or to ride a broomstick "horse." You can encourage them to play with dolls or animals and to be the individual or the doctor. They may also have strong attachment to a special toy or blanket that they will want with them when they are going to bed, an appointment, or the hospital.

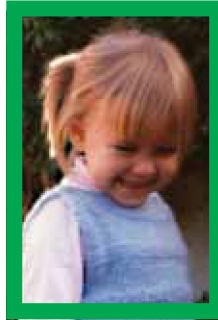
Piaget notes while their language skills increase and mature at this age, they cannot think in a concrete logical way (though they will be learning how to argue with you!). Many times their reasoning is magical not logical. They tend to see the world only through their own point of view (egocentric) and lack the ability to see it from anyone else's.<sup>[21]</sup>

An important part of parenting any child is of course instilling compassion, respect, and concern for others. Your expectations of behavior should be no different for your child with cystinosis.

## Explaining Cystinosis to Preschoolers

To explain cystinosis to a preschooler, it is best to keep it simple, age-appropriate for the child's cognitive level, and honest. Children need to know they did nothing to cause cystinosis, but also that it will not go away. They need to know the medicines will help them feel better and keep them from getting sicker. They can be shown where their kidneys are and how kidneys help clean the blood (since they will be hearing about the kidneys at medical visits). They can understand how good nutrition (however they get it) is important to help them grow and be healthy. Siblings or playmates need to know that they cannot "catch" it like a cold or the flu.

## EXPLAINING CYSTINOSIS: SERENA



*Serena, age 4.*

We were honest with all family and friends at the time of our daughter's diagnosis. The doctors told us that children with this condition didn't live long. They told us to go home and enjoy the time we had left with her. She was 18 months old. We knew little about cystinosis back then, and there was no support.

We had an older child who was five years old when Serena was diagnosed and we were always open and honest and discussed things with the kids each step of the way. I remember meeting a child with cancer at the kids' hospital. When I chatted to the parent later they said the child doesn't know they have anything wrong with them. I remember thinking "you have to be joking." That child knew he was sick. They understand more than we give them credit for sometimes.



*Serena, age 30. Serena was seven years old when she received her kidney transplant from her mother.*

Honesty is the best way to go. If you try to hide things they know it and I believe you will lose their trust. Things can be explained simply when they are little. Something along the lines of: you have this condition called cystinosis and you have to have special medicine that other kids don't have. This is to keep you healthy. Some of them you will hate, some taste and smell terrible but if you don't have these medicines (or injections) you will get really sick and could die. We want you to stay as healthy as possible. We really wish you didn't have to take all of this stuff but there are 2,000 other kids in the world with cystinosis who have to have the same medicines every day and we might be able to meet some of them if you would like.



## Medications, Nutrition, and G-tubes

The medications a child with cystinosis will need are many and may change over time. Oral cysteamine, manufactured as the drug Cystagon®, however, is one that will not change over time. Both pre and post-transplant, this drug is the key to help remove cystine from the cells of the body and slow the progression of the disease. Other medications will be used to replace electrolytes lost by the kidney as a result of Fanconi's syndrome or to treat anemia or bone disease as the kidney function decreases.<sup>[6]</sup> Most medications come in liquid form or can be dissolved easily to make it easier to administer to the young child. As the child grows and learns to swallow pills, taking medications becomes easier.

The use of a G-tube in a child with cystinosis is a question many parents may face. It is one that requires careful consideration with input from the parents and doctors to determine what best meets the needs of the individual child.

Poor nutritional intake, failure to thrive, frequent vomiting, electrolyte imbalances, large urine output, and insatiable thirst are hallmarks of cystinosis in the first few years. After diagnosis many vile tasting medications are added to the daily routine. For many parents getting enough food and medicine into their children with cystinosis to promote growth and manage the disease symptoms becomes a monumental struggle. At this time the use of a G-tube may be considered.

A G-tube or gastrostomy tube is a tube placed into a surgical opening (gastrostomy) through the abdominal wall directly into the stomach under a general anesthetic. Through it, liquid medications, and nutritional supplements such as Pediasure or Suplena can be delivered directly into the stomach without the child needing to swallow. This is done through a bolus-feeding syringe or a special pump that can deliver a specified amount of fluid every hour.

**Advantages:** Administering medications is much easier if the child refuses to take them by mouth. Caloric and nutritional intake can be increased with nutritional supplements to improve growth and nutrition.

**Disadvantages:** Inserting a G-tube is a surgical procedure that requires general anesthesia. The size of the tube will need to be changed as the child grows. As with any surgery there is a risk of infection. Care must be taken not to become G-tube dependent for all nutritional intake.

A G-tube should not replace oral nutrition in the child with cystinosis. Supplements given through the tube are

just that - supplements to provide adequate nutrition and calories to support health and growth until the child can orally consume enough on his own. The child should be encouraged to eat every day, and to participate in meal times with family whenever possible. As the medications and supplements regulate electrolyte imbalances, usually the appetite improves.

The decision to use a G-tube should be based on the answer to this question. Will using a G-tube improve the quality of the child's life? <sup>[22], [23]</sup>

## Hydration and Toileting Issues

Dehydration for a young child with cystinosis is a constant concern. Cystine accumulation damages the kidneys ability to reabsorb water and various electrolytes causing large amounts of both to be lost in the urine. This is called Fanconi's syndrome. Cystinosis is the leading cause of Fanconi's syndrome in children.<sup>[6]</sup>

A child with cystinosis always needs access to fluids. A water source should be available at all times in all places, including home, daycare, preschool, in the car while traveling, church, doctors offices etc. Family, caregivers, and teachers must understand that fluids must never be restricted in a child with cystinosis.

Since these children also vomit frequently, this can also be a dehydration concern. Children with cystinosis may require hospitalization for IV rehydration more frequently than a normal child.

## Signs of Dehydration.<sup>[24], [25]</sup>

- Dry skin, tongue and lips
- Reduced or no tears, eyes that appear sunken in
- Fatigue, mood changes (fussiness or irritability)
- Poor skin turgor
- Rapid heart rate and low blood pressure

**Note: The most common signs of dehydration (thirst and low urine output) are NOT reliable indicators in children with cystinosis.**

Another issue is heat intolerance. Many children with cystinosis suffer heat intolerance if exposed to long periods outside in the hot sun. The cooling mechanism of sweating doesn't work normally in some people with cystinosis.<sup>[26]</sup> Avoid prolonged exposure to hot sun.

Toilet training in children with cystinosis may be delayed. This occurs because of the tremendous amount of urine that is produced each day by the kidneys. Children with cystinosis feel the same sensation of a full bladder as normal children, but may experience more "accidents" than a normal child. Parents can help by being proactive and planning ahead.

Always have extra sets of clothes, at school, in the car, or any place the child may be for any length of time. Build in frequent bathroom breaks into the routine of the day. Be sure to scope out the locations of public restrooms if away from home. If traveling any distance, keep urinals or bottles handy (admittedly this is easier for boys) or make frequent rest stops. Be sure that caregivers or teachers understand the need for unlimited access to water and the bathroom. They should clearly understand these children drink and urinate so much because their kidneys are damaged. They have no control over either.

A low key, no big deal attitude by caregivers, teachers and parents will help when the inevitable "accidents" occur to promote a positive self-esteem. Adults in the child's world should be aware of signs of bullying or teasing by other children regarding this issue and step in to stop it.

For many reasons staying dry at night may take longer. Almost since birth these children are used to being wet at night. Being soaked is such a normal state that many sleep right through cold, wet clothes and urine soaked sheets. As parents we've all tried diaper doublers, towels, incontinent pads, and changing sheets at night and washing them daily. It all becomes part of the new definition of "normal". Factor in additional fluid some of these children get through nighttime nutritional supplements and the problem is compounded.

Some parents of children with cystinosis have reduced bedwetting by making a routine of getting the child up regularly throughout the night to urinate, perhaps at the same time a medication is due. Other parents have had success with a bed wetting alarm system. A moisture sensitive pad attached to a wire and clipped to the child's underwear or pajamas sends a signal to a battery-powered

box that beeps loudly when moisture is detected. The noise is loud enough to wake up the child. In a very short time these children learn to awaken to the sensation of a full bladder before urination starts. They may wake up several times throughout the night, but wake up dry in the morning.<sup>[27]</sup>

From personal experience with our child this process took about six weeks. I was amazed how well it worked. Post-transplant bed wetting issues are fewer because urine volume is much less.

## COPING WITH MEDICAL VISITS AND HOSPITALIZATIONS

The new "normal" that becomes a part of life with cystinosis involves frequent doctors' office visits with pediatricians, nephrologists, and other specialists. It involves weekly or monthly blood tests and sometimes hospitalizations for dehydration, electrolyte imbalances, renal failure, infections, or other problems.

The stresses involved in this new "normal" vary from worries about the cost of healthcare (access to insurance, copays, out of pocket expenses), time off of work to handle medical issues, and childcare. These practical and financial concerns pile up on top of helping the child with cystinosis cope with the illness itself. It can quickly become overwhelming. Planning ahead can help decrease stress.

Learn everything necessary about your specific health insurance plan, including what is covered, what isn't, and which procedures may need prior authorization. If one knows what it's to do and what it's to cross ahead of time, the frustration may be lessened. Most states have supplemental insurance (Children's Special Health Care Services) that can help cover the cost of expensive medical treatment and medications for children with chronic disease. Access to and costs for state-provided insurance is based on income, and may cover what "private payer" health plans don't. Social workers at your medical center can assist you in learning about these options, and you can also search online to find information about your state's program by starting here: <http://www.insurekidsnow.gov/>

Talk with employers (carefully) about the situation and your need to take time off of work for doctors' visits or hospitalizations. While they may be empathetic, you may need to be prepared to make compromises with your work



schedule or work from home if your job allows. Some families find it works better to have one parent “stay at home” until things normalize. Each family needs to look at their budget, and their availability and flexibility around work and other family obligations. This may also help determine how parents divide up care tasks with each other or other caregivers (for example, late

night or early morning medications doses, or bed-changing and laundry duty). Finding time for these conversations can help you react more efficiently when crises happen, and prevent hidden resentments from building up over who is doing what.

Plan ahead with family or friends who can come in at a moment’s notice to help care for siblings, provide a meal, or do whatever else is required. In such situations friends and family want to help, but often don’t know how. To learn how to ask for help when needed, and then accept help with gratitude when it is offered is a gift for all involved.

## Dealing with Fears, Frustrations, and Mood Changes

Life with chronic illness can bring about fear, frustration and mood swings for your child with cystinosis, for parents and caregivers, and for siblings. Life can escalate to the point of implosion quickly.

The young child with cystinosis may not understand all the “sticks” of lab test, feeling sick all the time, being force fed vile medications five or six times a day, or being separated for periods of time from mom and dad during hospitalizations. They may react with tantrums and meltdowns that add further to the mountain of frustrations.

The easiest thing in the world is to cater to a sick child – trying to get them to eat – by offering bribes if they will swallow this dose or take just one more bite. It becomes easy to bend the rules for the sick child because parents feel sad for all the pain and suffering the child endures each day; but most of the time that just exacerbates the unwanted

behavior. All children need routine, clear expectations, and to know what the limits are.<sup>[28]</sup>

For the child, fear of the unknown can be the greatest fear of all. Try explaining what to expect in simple terms ahead of time. Be honest about the facts, explaining in terms he can understand. Be with him as much as possible during office visits, procedures, and hospitalizations. Bring a favorite toy, blanket or book from home to relieve anxiety. During hospitalizations utilize



Child Life programs available in many hospital settings, such as videos, games and playroom activities. These can provide a fun, positive distraction from less pleasant activities. Giving the child choices as much as possible in his treatment. (e.g. “Would you like apple juice or water with your medicine?”) will help him feel a little control over his life when so many things are out of his control.

## Developmental Issues and Starting School

Developmental delays are common in children with chronic illnesses and cystinosis. Poor nutrition, electrolyte imbalances, dehydration, kidney disease and frequent hospitalizations all combine to delay or impair normal physical, cognitive, or psychosocial growth in these children. Neurological research has shown children with cystinosis have significant deficits in visual processing, specifically visual spatial and visual motor skills that can lead to learning disabilities in math, spelling, or geography.<sup>[29]</sup> However, each child is different, so your child will have their own strengths and weaknesses, and their own learning style. Knowledge is power. Parents need to educate themselves about normal developmental stages to recognize any delays they may see in their children with cystinosis. Being proactive and seeking out early intervention programs can be crucial in addressing some of these issues.

Federal law mandates that children with medical conditions who have significant developmental delays are entitled access to Early Intervention programs through Part C of the Individuals with Disabilities Education Act (IDEA). The criteria for qualification to these programs may vary from state to state. The hope is that through early intervention the child's delays may be decreased, reducing the need for special education services later on.<sup>[30]</sup>

Parents who suspect a significant developmental delay in their child can request an evaluation through programs such as Project Find whose purpose is to identify developmental delays in young children and initiate strategies for interventions. More can be learned about these programs on special education web sites at the state level or through the individual school district web site.

Beginning at age three children, with special needs may qualify for special education services to address developmental delays following testing and the implementation of an IEP (individual education plan). Services may include speech/language, occupational therapy, physical therapy, social work or time with a special education teacher in a resource room or special education preschool setting.<sup>[30]</sup> Children with cystinosis may also

require special accommodations in the school setting for medication administration, fluid or bathroom access or other needs such as additional time to take tests. This may require having a 504 Plan in place. This refers to Section 504 of the Rehabilitation Act and the Americans with Disabilities Act, which requires accommodations be made available to meet a child's documented need in the school setting.<sup>[31]</sup>

Parents and educators working together as a team to meet the child's learning needs is ideal. Honest, frequent communication between parents and educators is the key. Parents can best help their children in the school setting by providing detailed information about cystinosis and the needs the child has as a result of the disease to the teacher or other staff involved in the care of the child. It is important that the child's educational needs are addressed in an IEP or accompanying 504 Plan. Information should be updated as needed and certainly yearly as the grade level and the teacher change. If absences occur due to illnesses, doctor visits, hospitalizations or lab tests, it is important to keep the school informed to assist the child in keeping up with missed lessons.<sup>[31]</sup>

## CHECKLISTS EARLY CHILDHOOD, AGES 3 - 6

### Parent/Caregiver Checklists Early Childhood Ages 3 - 6

- Learn about cystinosis, treatments and medications.
- Become familiar with all details of your health insurance plan and any additional supplemental plans available
- Educate others in the child's life, such as extended family, childcare providers, and school personnel, about cystinosis and the child's needs as a result.
- Talk to the child about cystinosis in an honest, age appropriate manner.
- Maintain normal routines as much as possible to foster normal psychosocial growth.
- Provide the child opportunities for choice in his care as much as possible. (e.g. "Do you want water or apple juice with your meds.")
- Keep a personal medical record for the child including medications with a time/dose schedule, labs, pediatricians, and specialists involved, insurance information, and contact information for all.

- Learn about normal physical, cognitive, and emotional, development in children to become aware of delays in the child with cystinosis that could impact educational opportunities; then act to address these delays early.
- Address toileting issues individually, proactively and realistically until continence is achieved.
- Recognize signs of stress in self or others and seek help when needed from friends, family or professionals.

### Child Checklists Early Childhood Ages 3 - 6

- Describe cystinosis to others in simple terms.
- Learn more detail as age and cognitive ability increase.
- Cooperate with medicines and therapies.
- Begin to identify symptoms, and tell an adult when something doesn't "feel right."



## SCHOOL – AGE, AGES 6 - 12

Piaget theorized that children who are ages 6 -12 are in the concrete operational stage. In this stage they have difficulty understanding abstract terms such as freedom. Children understand literal and concrete subjects. A sense of fairness is also developing.<sup>[32]</sup>

According to Erikson, a child at this stage has a desire to accomplish and master new skills. The focus is on mastering tasks, which are essential for success in adulthood – reading, writing, math as well as interpersonal communication skills and physical skills. The focus is on education and competition.<sup>[33]</sup>

### Helping Your Child Understand Cystinosis and Treatments and Talking to Others

**Knowledge Is Power.** As children grow up, their ability to understand information and assume responsibility for their own care increases. Every year or so, a parent should ask your child what they understand about cystinosis and then fill in any gaps and correct misperceptions.

**Older School-aged Children** are more capable of understanding cystinosis and its treatment, but they should not be expected to react as adults do. Parents may feel the need to protect their children by restricting them from activities with other children. This is a natural reaction, but it can interfere with the child's independence and sense of mastery. To the extent allowed by the child's doctors, parents should help the child to participate in school or other activities.

Information and support can be empowering. Meeting other children with cystinosis can make a child feel less alone. The Cystinosis Research Network Family Conferences are an opportunity for children to form life long relationships with others who are experiencing similar challenges. There are also websites specifically for children with chronic illnesses.

- **Kidshealth.org** has kid-friendly information on all kinds of health and illness topics.
- **Bandaides and Blackboards for Kids**, is a site for kids with chronic illnesses or other medical problems.

### Coping with Hospitalizations and Illness

Hospitalizations and illnesses may delay goals from being obtained. A child may miss school, birthday parties, piano recitals, or soccer games due to complications of cystinosis and this can lead to a feeling of frustration and sadness of being cheated out of experiences that their peers are having.

### Helping Your Child Cope

Children with cystinosis often deal with more stress than their peers. For example, they may have to cope with medication side effects, frequent medical appointments and hospitalizations, painful injections, or surgery.

Unfortunately, there are no simple ways to help your child avoid these stresses. Here are some suggestions that may make the situation a little easier.

- **Listen to your child.** Whether she is feeling sadness or frustration, it is helpful for her to express her emotions. She should feel that she can share her thoughts and fears without your overreacting or becoming upset. Ask how she is feeling. Be available and supportive. Listen not only to what your child says, but also try to hear what is left unspoken.
- **Inform your child about what lies ahead.** Anxiety is often based on the unknown or on inaccurate presumptions about the future. Find out what your child does and does not know. Explain exactly what will happen during an upcoming doctor's appointment or hospital visit; if you are unable to answer all your child's questions, both of you should talk to the doctor. Do not expose a child to a frightening procedure unless she has been informed of it beforehand. Conversations with other children who have gone through the same experiences can be invaluable.
- **Frequently talk about cystinosis** so that your child feels comfortable being open about it.
- **Emphasize your child's strengths** – the things she can do well despite cystinosis.
- **Help your child feel that she can be in control of some aspects of her situation.** Try to find choices that can be given to her, such as which arm to have blood drawn from, when a procedure will occur, or what reward she will get for cooperating.

## School Issues

Understanding cystinosis will help your school system understand your child or teen. The school may not have the intimate understanding that parents do, but having a clear and accurate idea of the impact of the illness will help the school and district serve your child's needs.

Parents should not be intimidated by the “educational professionals”, and the school administrators should NOT be intimidated by parents! Families should be clear, calm, and diplomatic in interactions with school and district staff. It is best to work as a team. However, parents should not let teachers, support staff, or administrators tell them that a request is “impossible.” An individual with cystinosis has a right to have his or her educational needs met.

Some (but not all) children with cystinosis may have particular learning problems with visual processing, math, and spelling<sup>[30], [34]</sup> There are alternate ways to teach almost everything! Some children will need full evaluations (Individualized Education Programs or IEPs) for special education services; some will need tutoring or more time to finish tasks. Some will need 504 plans (a documented need for special accommodations in accordance with Section 504 of the Rehabilitation Act and the Americans with Disabilities Act) so they can receive medications in school, or have access to water or the bathroom, when they need it.

## Communication Is Key

Parents should get to know key individuals – including a child's principal, guidance counselor, nurse, psychologist, social worker, AND ALL teachers (including those who teach special classes like art and physical education) as soon as the school year begins. The principal should be asked to designate a “point person” within the school as someone who can be approached when questions arise. For medical issues, this will likely be a nurse. For academic issues, it will likely be a guidance counselor or school social worker. Keep in mind that some teachers may be more helpful than others.

Provide the school with a packet of information about cystinosis. There are numerous helpful resources, pamphlets, and articles at: <http://www.cystinosis.org>

If a child is having cognitive or learning difficulties, parents should ask for a school evaluation, but also speak with medical experts about what might be happening? If a child with cystinosis is not able to attend school for an extended period of time due to health issues, there are options for quality home instruction (though it is often advisable to keep it as short as possible) There may be technology that will allow a student to be in the classroom “virtually” during extended periods of absence.

## CHECKLISTS FOR SCHOOL AGE CHILDREN, AGES 6 - 12

### Parent/Caregiver Checklists School Age Children Ages 6 - 12

- Meet with the school staff in the spring of every school year to review your child's IEP and 504 plans and implement any necessary changes.
- Meet with the school nurse and staff at the start of the school year. Bring handouts from the CRN website and discuss your child's individual needs for water, frequent bathroom breaks, medications, and eye drops.
- Help your child to become more independent with self-care. Just as they learn to do chores around the house during the school-age years, your child should also start to learn how to care for themselves. Have your child help prepare weekly / daily medications. Discuss the purpose of each medication with your child. If parents can help their child to grow toward complete self-care as a process over several years, it can be an easier transition

for the adolescent or young adult to completely care for themselves when going to college or moving out and getting a job or starting a career.

### Child Checklists School Age Children, Ages 6 - 12

School age years are all about learning and speaking up for oneself. In addition to the preschool expectations, the school age child should be expected to:

- Know the dose, time and correct use of medications.
- With the help of an adult, seek ways to remember medicines. For example, have a daily routine, such as taking morning medications with breakfast, and evening medications with dinner and/or at bedtime.
- Discuss treatment of symptoms with parents.
- Learn to quickly identify signs that you are not feeling well.

## Building Bridges

- Focus on creating confidence, resilience, and building strong communication skills.
- Set standards for independence.
- Having a disease such as cystinosis can alter a child's self-confidence and self-esteem. It is important that the child feels good about themselves as a person and is proud of who they are.

## Building Self Confidence

Self-confidence means having a positive and realistic opinion of yourself and being able to accurately measure your abilities. Having cystinosis can and sometimes does interfere with your child's ability to participate in activities as their peers do. Self-confidence can help your child to think positively and deal better with the daily stresses of having cystinosis. But remember, some of the self-doubt of adolescence helps teens figure out who they are and what they care about. Below are several ways to help your child build self-confidence:

- Recognize your child for doing a good deed, doing well at school, or completing a goal.
- Point out their strengths.
- Spend quality time with your child on an activity they enjoy (even if it's not YOUR favorite!)
- Applaud your child's effort to improve.
- Practice positive reinforcement.
- Listen to your child when he or she speaks. Do not ask them yes / no questions. Ask them open-ended questions and give them time to answer. Let them know they are being heard.

## LIVING WITH CYSTINOSIS: KACY'S STORY



Kacy was considered a late diagnosis at four years old. She certainly had symptoms: including failure to thrive (beginning around 11 months), extreme urination and thirst, constipation, and protein in her urine (occasionally). But she never suffered from any

gastrointestinal issues, ate normally, and had the energy of any normal four year old. We were having her tested for growth hormone deficiency, which required a 12-hour fast, which sent her into severe dehydration and a two-week hospital stay. That event, and the eye exam, led to her being diagnosed with cystinosis.

The most difficult time for us was during the first year. She came home from the hospital with a G-tube and five different liquid medications. We hydrated her overnight with a pump and managed to find a new normal. She never really blinked an eye. She went back to her normal activity and energy level, returned to preschool, and her health stabilized quite quickly. She began to grow on a normal pattern, although far below her age group because of the late diagnosis. During the spring of her eighth year we switched her over quite easily to all oral medications and in the fall of that same year she requested to have her G-tube removed. It has been a year since that occurred. We only get nervous when she contracts the stomach flu, which she has twice since her G-tube was removed. Both times we managed her care at home with no hospitalization.

A typical day for Kacy starts at 7 am with a wakeup call and ½ of her morning medications (six pills). She usually goes back to sleep for an hour and wakes for the day at 8 am, finishes her morning meds (7 pills) and an eye drop before she heads to school at 9am. At school she gives herself 3 eye drops at 10 am, 12 pm, and 2 pm. At 2 pm she also takes eight pills. She comes home at 4 pm, takes two iron pills and another eye drop. At 8 pm she takes her evening medications (12 pills), another eye drop and her growth hormone injection. Bedtime usually comes by 9 pm.

Kacy swims competitively three days per week for an hour and dances two days a week for an hour. Swim meets follow

on the weekends and many play dates with friends. She eats normally at every meal with no side effects from any of her medications. Occasionally she will vomit in the mornings when her pills go in on an empty stomach, but those times are few and far between now. We are thankful for that. She is growing well on the hormone shots (12 inches in the last couple years). She has now reached the average height of girls her age. We know that a kidney transplant is in her future, but for now her kidney function is roughly 66%. She sees her nephrologist and ophthalmologist every 3 – 4 months.

## LIVING WITH CYSTINOSIS: SARA'S STORY

On June 22, 2005 Sarah entered the world as a perfect, chubby, “normal” baby. She was the largest of our four children, full term, and the result of a thankfully uneventful pregnancy.

At 12 months of age, Sarah stopped growing, eating well, and sleeping well. At 13 months of age, she began frequent vomiting, extreme thirst for water only, and very heavy urination. Frequent trips to the doctor resulted in disappointment and frustration. “She is just small.” “She is lactose intolerant.” “She is a picky eater.” At 14 months of age, Sarah vomited twice and was totally dehydrated and the doctor was “too busy to see her today”. Right then I got a new pediatrician. Sarah was admitted to the hospital for fluids and many tests. Within 24 hours, blood work identified renal tubular acidosis: a malfunction of the kidneys caused by Fanconi’s syndrome. A pediatric nephrologist was called to see our tiny, very sick child. He sat us down and explained that he thought Sarah had cystinosis, which was causing the Fanconi’s syndrome. We were devastated but determined to fight to save our daughter. The test for cystinosis took two weeks to be delivered to the hospital. Blood was drawn and sent back to the lab in California. After two more agonizing weeks, cystinosis was confirmed.

Sarah quickly encountered many challenges in the process of trying to stabilize. She had rickets, failure to thrive and cystine crystals in the corneas of her eyes. She needed physical,



occupational, and speech and development therapy to help her catch up. By 18 months of age she had a G-tube and was taking medicine around the clock. Blood draws became routine. We had doctor appointments with the pediatrician, nephrologist, dietician, ophthalmologist, surgeon, and endocrinologist to go along with the weekly therapy sessions. This was a whole new level of busy for our family!

Sarah is six years old now and in kindergarten. She loves school, has many friends and loves to play dress up and paint her toes. She rides horses, goes to the beach, and loves everything pink. She also takes 27 pills, one injection, and 14 eye drops a day. She has supplemental feedings through a tube. She has conquered rickets, failure to thrive, and most of the crystals in her eyes. She has graduated from all the therapies and only has blood work once a month. Sarah is funny, active, smart, and kind. She also is woken up every night for medicine, is not potty trained at night, does not sweat, and rarely picks up her toys! The vomiting has greatly decreased and Sarah is growing. She still visits all her doctors, which now include an ear nose and throat doctor and a gastroenterologist. She thinks of herself as an ordinary kid. We think she is extraordinary.

## EARLY ADOLESCENCE, AGES 12 – 15

Younger teenagers (ages 12 - 15) generally have some behaviors in common<sup>[35]</sup>, whether they have cystinosis or not. They will likely:

- Alternate between acting like a younger child and like an older teen
- Struggle with a sense of identity
- Experience moodiness.
- Worry about being “normal”
- Begin to test rules and limits
- Express feelings by actions rather than words
- Begin to place more importance on friendship
- Pay less attention to parents
- Begin to notice and react to peer pressure
- Find new people to love and feel physical attraction
- Gravitate toward same-sex and mixed-gender group activities



## Taking Medications and Taking Responsibility

Young teens should recognize what their medications look like and should be able to tell their parents and healthcare team the following:

- Name of each medication
- Dosage and schedule for taking each medication
- Purpose of each medication
- Side effects of each medication

Young teens are typically accustomed to their parents reminding them to take their medications. They should be exploring ways to remember to take their medications on schedule. Parents can assist their children in finding a method that works, such as setting an alarm on a watch, sending texts, or setting reminders on cell phones.

## Coping with Medication Side Effects

Children with cystinosis may require many medications in order to manage their disease. Each medication comes with its own set of side effects. Managing these side effects can be stressful and often times discouraging. Occasionally, medications are added to the already complicated medication regimen to offset the side effects of the treatments used to manage cystinosis! Side effects that are really bothersome can lead to a teen avoiding vital medications. It is important to have an open line of communication early about side effects and strategize together – before cystine levels rise or a kidney transplant is endangered. If the side effects of a particular medication are unmanageable, speak with the healthcare provider to see if alternate medications are available.

## Teach Teens to Self-Advocate

All of us need to learn to communicate what is important to us, and it's never too soon for parents to start teaching their child to be assertive. People with cystinosis will be placed in many circumstances where the need to advocate for themselves will be crucial. Adolescents with cystinosis should feel comfortable with speaking up for themselves. They should be able to express their needs and preferences at this age. Parents should encourage children to practice self-advocacy while still supporting them in their decisions. It may

be helpful to role-play scenarios that may come up at school and with friends to help young teens gain self-confidence in advocating for themselves.

Bullying stinks! Children with chronic illnesses may be hidden victims of bullying. Parents are a child's number one advocate in this. Even when the lines of communication are kept open, children still may be afraid to approach parents or teachers for help in dealing with a bully. Children may fear retribution for getting the bully in trouble or feel embarrassed or ashamed. Parents should stay involved and supportive of their child's school and extra-curricular activities and remain aware of what the child is doing and whom he or she is spending time with. Teasing and isolation should not be accepted! Teens should decide with whom they want to share information about cystinosis (close friends, teachers) and what they want to keep private, especially if they are entering middle school or high school in a new place.

### Watch for These Potential Bullying Warning Signs\*:

- Loss of interest in school and extracurricular activities
- Frequent complaints of illness to avoid attending school
- Sudden decrease in academic performance
- Few or no friends with whom he/she spends time with
- Unexplained bruises, scratches, and cuts
- Fear of going to school, riding the bus, or walking to school
- A preference for a long or illogical route to school
- Increased moodiness, sadness, or depression
- Loss of appetite
- Trouble sleeping
- Anxiety or low self-esteem

\*Adapted from [mychildsafety.net](http://mychildsafety.net)<sup>[6]</sup>

## School Issues and Transplant

Honesty is the best policy. If transplant is near, if there are particular medical concerns, or if a child is starting in a new school, a meeting should be held with all of the key people at the beginning of the year or semester. Again, provide a packet of information to everyone. Not sharing information does not allow understanding, and can create problems.

There will be many lab visits and appointments post-transplant. Make sure that it is clear to teachers and school staff before transplant that protecting the new transplant and avoiding infection will be the first priority for a while. There will be absences (do be sure to document visits or hospital stays). Let the school know to not hold it against the student when there are frequent medical absences.

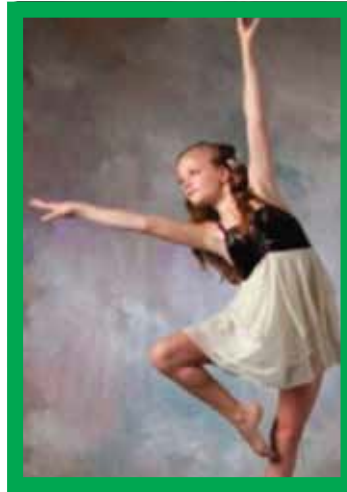
If a young teen is using illness as an excuse to miss school, or telling school that he or she is ill when it is not the case, parents face a challenge. It is advisable to address it as parents might address other kinds of non-adherence. Try to understand what is happening. Support rather than punish and involve the teen in finding a plan that works. NOT going to school is NOT an option.

Encourage school staff to ask questions. Tell them they can leave voice mail, send e-mail, or text message anytime. Urge them to ask questions, both academic and medical. There are no dumb questions! Make it personal – about the individual – and make it a good learning experience for school staff. They can be your advocates!

## Friends

Encourage friends to reach out if a child with cystinosis is out of school for more than a few days. Friends may ask how they can help. It is important for teens to stay in communication with their friends, be it through cards, e-mails, texts, phone calls, or Facebook. Staying connected will make returning to school – after days, weeks, or even months – much easier.

## LIVING WITH CYSTINOSIS: LAURA'S STORY



Laura's first few years after diagnosis were rough. She averaged a two pound weight gain for her first five years, which if you do the math leaves for one tiny little person. Fanconi's syndrome was extremely hard to control and we had to get creative to get her into some sort of normal range. She had no "reserve" and her asthma added into this equation and made for

many hospital trips. Vomiting was a way of life for us. Laura could "toss her cookies" into any container. We found that tubs like the large ones that cookie mix comes in worked great, as they had a lid so you could cover it when the fun was done. To this day we still keep gallon zip lock bags in our cars because you just never know.

Two years ago, Laura had one leg operated on to help with her "knocked" knees. She has responded well and has corrected nicely and will have her "8" plate removed this summer. Her G-tube was removed when she was 12 and the look on her face when she saw her stomach without a tube is one of the most precious moments of my life. She rubbed it just like she had as a baby. It still brings a tear to my eye thinking of how beautiful she thought her stomach was. We tried growth hormone on two separate occasions but for Laura it was not worth the trauma. She is "needle phobic" I say that with a grin, since you can prod her veins all day, but try and give her a shot and she goes ballistic. Nothing will persuade her to willingly get a shot, and we are firm believers in picking your battles. Laura is well aware that she is going to be a small woman and she does not care. She is 4' 8"ish, 80 pounds and quite happy with herself.

A day in the life of Laura depends on the day; no two are ever the same. She is on intermittent homebound service, which means she goes to school when she feels up to it; otherwise she has a teacher come to the house who brings her assignments and helps her as needed. This has been her school structure since 7th grade and she is currently in the 9th. She has a restricted license, which allows her to drive

alone until 6pm, which has done wonders for her self-esteem and both her and my mental health since we get time away from each other. She dances competitively in jazz and hip-hop, which she loves. She has friends who have been close for years and she spends at least one night a week with them hanging out or on sleepovers. She has many friends who are older who have known her throughout her life. They are like big sisters to her and she loves to spend time with them. Our standing joke for years was that she liked her friends to have cars so they could take her places, but now that she can drive we have to admit she likes her friends because they are great girls. Our life is not completely “typical,” but we have made a happy life and fill it with as much love and joy as we can.

## BUILDING BRIDGES: BUILDING SELF - CONFIDENCE

Self-confidence means having a positive and realistic opinion of yourself, and also being able to accurately measure your abilities. It is important that teens feel good about themselves and are proud of who they are. Having cystinosis can and sometimes does interfere with a child's ability to participate in activities as his or her peers do. Having a disease such as cystinosis can alter a child's self-confidence and self-esteem.

Self-confidence can help a teen to think positively and deal better with the daily stresses of having cystinosis. But remember, some of the self-doubt of adolescence helps teens figure out who they are and what they care about

**Below are several ways parents can help their teen build self-confidence:**

- Set standards for independence
- Focus on building confidence, resilience, and strong communication skills
- Recognize their child for doing a good deed, doing well at school, or completing a goal
- Point out their child's strengths
- Spend quality time with their child on an activity he or she enjoys (even if it's not parent's favorite!)
- Applaud their child's effort to improve
- Practice positive reinforcement

Listen to their teen when he or she speaks and use open-ended questions rather than ones that require only a yes/no response

## CHECKLIST AND EXERCISES

This is a time when young teens can begin to talk directly with their healthcare team. They should be encouraged to spend a few minutes talking privately with the team at the beginning or end of each visit.

Young teens can sit in (or pick up the phone) when parents are making medical appointments or calling for prescription refills. Parents should model for their children how to interact with health professionals to receive or provide information.

Families should start discussing hopes and plans for after high school (even if they seem unrealistic or far away). Talk about how health concerns and college/career choices impact each other. Teens should be made aware of examples of people with cystinosis or with kidney disease or other chronic illness who have excelled.

Parents should ask the healthcare team how the transition process is handled. Then, the discussion can be repeated with the teenage individual present, so he/she can become familiar with the process as well.

### Sample Questions to Ask Your Medical Team

- At what age does transition happen at this hospital?
- Are there teen or young adult specific clinics?
- Who will help coordinate transition?
- Do you have adult specialists who you recommend?  
Why do you recommend them over others?
- Where are specialists located? How will we meet them?  
Do we need referrals?
- Are there adult physicians or specialists who are knowledgeable about cystinosis?

## OLDER ADOLESCENTS, AGES 15 - 18

Older teenagers, ages 15 -18, generally have some behaviors in common,<sup>[35]</sup> whether they have cystinosis or not. They will likely:

- Complain that parents interfere with independence
- Worry about their appearance and body image
- Pull away from parents emotionally
- Put more energy into maintaining and starting new friendships
- Adopt a strong “group” identity or identify with a certain “clique”
- Grieve the psychological “loss” of their parents, even while rebuffing parents’ attention
- Keep experiences private (and prefer keeping diaries, journals, or private blogs)
- Begin to value intellectual qualities and moral values and think about how their behavior impacts others
- Develop a sense of tenderness towards people they are physically attracted to
- Start to experience passion and love

### Moving Towards Independence

The transition years between being a teenager and becoming an adult can present difficult challenges. Young adults with cystinosis are confronted with a unique set of stressors in addition to the normal stress that teenagers face every day. Cystinosis can interfere with a teen’s comfort in becoming independent, and parents may be resistant to a teen’s efforts to be independent.

- The goal is to provide young adults with cystinosis with education that will encourage a successful and healthy future. A parent’s job is to provide information and resources to help adolescents think and live as independent adults.
- Parents cannot hold their child’s hand forever, even if it is their heart’s desire!
- Seek a balance between “normal” adolescence and the unique healthcare needs of having cystinosis.

## BUILDING SKILLS – TIPS FOR PARENTS



- Involve teens in ALL health-related discussions (treatment choices and current concerns about their illness).
- Teach teens self-care skills related to their illness (from medications to calling the pharmacy or scheduling doctor’s appointments). Parents should have their older teens sit in on phone calls, and then let them make the call themselves with adult supervision.
- Encourage teens to monitor and manage their treatment needs as much as possible. They can fill out flow sheets of meds or dialysis, or keep a notebook.
- Encourage the development of coping skills to address problems or concerns that might arise related to their illness such as:
  - Talking to friends about cystinosis
  - Participating in support groups
  - Expressing frustration or anger when needed
  - Using humor to defuse frustration or anger
  - Researching a problem
  - Participating in religious or social support activities
- Remember that the way parents cope sets an example for children.
- Encourage use of problem-solving skills related to their illness. Parents and teens can role-play and ask each other questions such as, “What do you think you would you do if...?” or “What do you think would happen if...?”



## Taking (and Not Taking) Medications

### For Teens

- Think about how much privacy is important to you at medication time. (See next column.)
- Find a pill case, small sack, or cool case for carrying daily medications and eye drops that can be tucked in a backpack or pocket during social activities.
- Speak honestly with your parents or your healthcare team if you are having a hard time taking medications – whether because of side effects, a busy schedule, or sheer frustration. There may be options that could make things easier.

### For Parents

- Many teens will be taking medications both to control cystinosis and to protect a kidney transplant. The reality of getting a kidney transplant in adolescence can be overwhelming.
- When a teen's chronic illness reaches an unstable state due to “non-adherence with treatment recommendations” (such as not taking medications as needed), aim for discussion of what's happening rather than arguing or punishment. Decrease your and the healthcare team's frustration and replace it with support.
- Be honest about the results of not adhering to medication schedules, but try to avoid scare tactics. Long-term outcomes may not make sense to teens. Focus on how non-adherence or poor self-management can affect daily activities such as going to school, playing sports, and learning to drive.
- Talk about what the non-adherence is really about – taking meds can be a burden, it may interfere with social activities, the side effects may be too troublesome, etc.
- Not taking medications can also be a way of “acting out,” which other teens might express with different troublesome activities (skipping school, drugs, alcohol). It can be a way of expressing rebellion...or a way of trying to feel in control. Work together as a family and with healthcare professionals to find a treatment plan that works for the young adult individual.

## Maintaining Privacy While Taking Medications

Some teens prefer absolute privacy while taking medications (in the nurse's bathroom at school) while others are comfortable being very public. Plan with school staff where medications will be stored, and know school system regulations about students carrying medications.

At home, try to keep medication time low-key. Keep it public if you can – in the kitchen at breakfast or dinner. A parent's knowledge that a dose is taken may help avoid conflict later.

## Coping with the Smell and Side Effects of Cysteamine

Cysteamine is THE lifeline for individuals with cystinosis. As children approach adolescence and young adulthood, it can be tempting to skip cysteamine due to social concerns, self-conscious feelings about the drug's odor, and complications such as gastrointestinal distress.

Without cysteamine, cystinosis will take its natural course, and more complications of the disease can happen at an accelerated rate. It may take cystine a while to build up in the system, so young teens and adults may not be immediately aware of the damage to their health. The issue of odor and ways to decrease the adverse side effects of cysteamine should be discussed and managed, so that a young person is not tempted to skip or stop taking this vital medication.

Methods used to deal with the smell of cysteamine include:

- Vitamin B2 (Riboflavin), either isolated or as part of a Vitamin B-Complex
- Chlorophyll
- Breath sprays and body sprays

Planning ahead and talking openly with trusted family, friends, or healthcare providers, while discussing worries and possible solutions, will ensure that adolescents with cystinosis do not feel the need to either withdraw socially or stop taking cysteamine.

## For Teens

Being a teenager is not easy – for many reasons. If a teen or young adult has five or more of these experiences, he/she may need more intense and immediate support than just talking with a friend or family member:

- Feeling sad all the time
- Getting angry easily
- Losing interest in activities that were formerly enjoyable
- Not eating at all or eating too much
- Sleeping too much or too little
- Missing school frequently or experiencing a significant drop in grades
- Worrying about being rejected or not doing something well
- Feeling too tired to do anything
- Feeling anxious and shaky
- Feeling worthless or guilty
- Feeling isolated; avoiding or not having friends
- Having problems concentrating
- Thinking about running away from home
- Thinking about self-harm or suicide
- Deliberately skipping medications.

These are symptoms of depression<sup>[37]</sup>, which can be treated in many ways, including individual, family, or group “talk” therapy. Some forms of depression might even be treated with medication. A doctor should be alerted if an individual is feeling this way. Having these feelings doesn’t mean a person is “abnormal” or crazy, but it does mean that he or she may be having difficulty coping.

## For Parents

Talk to your healthcare team and seek help if:

- A teen seems overwhelmed with emotional issues related to living with a chronic illness. This could take the form of extreme sadness/depression, anger, or isolation/withdrawal
- A teen’s development “regresses” or seems overly dependent
- A teen withdraws from or gives up interest in his or her usual activities
- A pattern of “non-adherence” continues, or risky health consequences occur. Not taking cysteamine can result in development of cystine buildup in all organs over time.

Not taking immunosuppressants can result in acute rejection of a kidney very quickly, or cause a slow decline in kidney function due to chronic rejection. When you have a chronic illness, sabotaging your health can become a powerful way to get attention or act out.



## For Teens

- Spend a day or weekend independently preparing and taking medications and eye drops – without parents reminding you! Set up your own medication box for the week.
- Got your driver's permit or license? Drive to your appointments. Otherwise, help the environment and figure out how to use public transportation to get there!
- Start exploring academic and work interests that you want to pursue after high school. Look for volunteer activities and try things out. Talk to friends or family members about their choices.
- Keep a journal or write when you feel strongly about something. Write an essay about your experience with cystinosis. Write about the best and most challenging things you have experienced (which might have nothing to do with cystinosis!). Writing can be a good way to understand who you are and who you want to become. Practicing and trying out different ways to tell "your" story may be useful for college and scholarship applications, or enable you to be an advocate for a cause you care about.

## For Parents

- Start talking to teens about how they can communicate with medical providers on their own. If they have a complaint or concern, they can contact provider independently, or with parent nearby.
- Start talking in-depth about plans for after high school and how career choice or school may impact your child's health.
- Start talking about transition to adult specialists. Consider finding a primary care doctor to coordinate the older adolescent's care.
- Find opportunities to meet adult care providers before the transition occurs. Some programs have transitioning clinics; if not, ask your physician about your hospital's transition protocol.

### Take the "Know Your Cystinosis" Quiz:

- When was your cystinosis diagnosed?
- When did you receive dialysis and/or transplant?
- What are your medications?  
What are they each for?  
When do you take them?  
Any side effects?
- Are you allergic to anything?
- Any major surgeries or hospitalizations?
- Who are your doctors and how do you reach them?

Knowing the answers to these questions is important as you prepare to transition to a new medical team. They are also vital to know if you are involved in after-school activities, out with friends, or traveling. In an emergency situation, like a sudden illness episode, a sports injury, or a car accident, you must be able to tell medical personnel about your health and your medications.

## CHECKLIST OF TASKS AND EXPECTED AGES

Some teens (and families) will be ready to tackle these tasks early, while others may choose to wait. These are suggestions of what teenagers may be ready for, cognitively (what they can understand) and practically. <sup>[38]</sup>

Task	Expected Age
Name and describe cystinosis	14
Name symptoms for cystinosis, transplant rejection, or other condition(s)	14
Have a basic understanding of genetics related to cystinosis	14
Discuss how cystinosis may affect the future	14
Take medications as prescribed	14
Carry a back-up supply of medicines and an updated list of medications	15
Explain consequences of not taking medications	15
List medications, dosage, and timing	15
Describe medication side effects	15
Go to all scheduled clinic visits and arrive on time, perhaps even traveling independently and meeting parent(s) there	16
Have a primary care doctor and know how to reach him/her	16
Know why it is important to avoid tobacco, alcohol, and illegal drugs	16
Know about sexuality, safe-sex practices, and the use of condoms to prevent sexually transmitted diseases – and be able to talk with parents, providers, and/or potential partners about these issues	16
Know about reproductive health issues related to cystinosis	17
Have a medical team that addresses all questions directly to the individual	17
See medical team independently – the team can provide parents and individuals with goals to be accomplished prior to visits to allow for independent interviewing	17
Get labs as scheduled and know what is needed when, and why.	17
Have awareness of insurance information (i.e. provider, changes in insurance at what ages, etc.)	17
Have a plan for school or work after high school graduation	17-18
Graduate from high school or receive a GED	17-18
Contact clinic to schedule appointments	18
Call for medicine refills independently	18

Checklist adapted from "Moving On – Transitioning from the Pediatric to Adult Health Care Team", University of Wisconsin Children's Hospital.<sup>[38]</sup>

## LIVING WITH CYSTINOSIS: GARRETT'S STORY



Hi, I'm Garrett, 16 and am here to talk about life as a teenager with cystinosis.

Right now I take 85 pills every day because I'm participating in a drug trial for the new extended release Cysteamine Bitartrate. I no longer have to get up in the middle of the night. Boy did that make my life better. To be 14 and finally get a night's sleep without interruption helped me in so many ways. Seems like a large number of pills, but I'm a big person so my dosages may be higher than many of yours. I work out and have a lot of muscles. My parents worked very hard to convince the doctors that growth hormone was appropriate for me, and I took a shot every day for 7+ years. It was something that I wanted, and participated in. Can't say that was my favorite part of the day, but it just became normal for me. Near the end when it was obvious I was in the growth curve and around the normal ranges, the doctors and my parents gave me the option to quit the shots, but I wanted to keep them up for as long as they worked, and they left the decision up to me. I continued with them till my growth plates were closed, and there was no more growth to be had. My Mom says I've always been very compliant with

taking my medication. When I was a baby, she would talk to me and say things like, "it is the rules", or, "you have to take your meds." My Mom and Dad always had a set or two of pill cases with them so we could always go out to eat or make plans on the go. My kidney function is still really good.

I'm a freshman in high school. I just lettered in a sport. I was on the rifle team and as a freshman earned a letter. My parents keep telling me this is a big deal. I haven't been active in a lot of sports because I don't sweat adequately. This sport however isn't outside and doesn't require running (which I love to do). I tend to overheat when I run or do other real physical activities. I have taken Tae Kwon Do, won awards in Go Kart racing, and love to swim. My favorite things to do are read, watch TV, play Xbox – just normal kid stuff.

My perspective on cystinosis is that I have gotten to go places because of Family Conferences (Detroit, Waterville, Orlando, Salt Lake City, San Antonio, San Francisco) that otherwise I probably would not get to visit. I know other people all over the world that I to see every two years, or sometimes more often. I've been involved in research studies in San Diego, San Antonio, and Atlanta. That means I'm involved in improving my own health, but also that of others. I get my own meds ready and most of the time I remember to take them on time. I know what my last cystine, creatinine and BUN levels are.

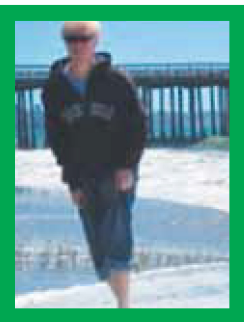
I think this disease is harder on my parents than me. For me everything is "normal". I've never known any different. I'm a pretty normal kid in my mind. Yeah, I take meds, have doctor visits and blood draws, but I've been doing it all my life. My two best life friends have been diagnosed within the last two years with life threatening diseases. One Lymphoma, the other Crohn's. Interesting that these two people who have accepted me for who I am all my life, now are dealing with their own issues. It's nice that I can be there for them and listen, and accept them as they have me.



## Transition to Young Adulthood and Beyond

The Cystinosis Research Network (CRN) created a **Cystinosis Pediatric to Adult Care Transition Guide**. The Guide is available on the CRN website at: <http://www.cystinosis.org>

### LIVING WITH CYSTINOSIS: STEVE'S STORY



I'm a 24-year-old college student majoring in Graphic Design and minoring in Marketing at Eastern Michigan University. Currently I am taking three art studio classes. I am enrolled in a marketing lecture class and I am heavily involved with the Ypsilanti and Ann Arbor music scene. All this keeps me very busy.

However, these things are not all I have to coordinate in my life. I also have cystinosis.

Having cystinosis thrown into the mix complicates everything in my life a bit. Medicine is easy for me take. I have alarms on my cell phone that remind me when to take my pills. The complicated part really is learning to function as an adult with a chronic disease. Because of my busy school schedule, my medical responsibilities that I manage myself and my responsibilities to others; I sometimes get burned out and need to get away from things for a while. The last couple months I've had a new medical issue come up along with a barrage of everyday life problems being thrown at me all at once. As a result I've had to learn how to reorganize myself, and deal with long standing issues in order to make peace with my life. For the most part I'm in a great place now. What I've been through has led to exciting and great new relationships I've never experienced, or expected to experience and these have helped me to discover a strength in myself I didn't know existed.

### LIVING WITH CYSTINOSIS: MACK'S STORY

I was born on April 9, 1964. My early years were normal, except for photophobia since I was 4 years old. At age 6, while in first grade, my slow growth became very noticeable. I was about a head shorter than other classmates at the time. It took a long time to determine a correct diagnosis in



the early 70s. Several doctors mis diagnosed my condition as diabetes. Our family doctor was determined to find somebody who could correctly diagnose me. When I was 8 years old, doctors suspected that I might have cystinosis. I was referred to doctors in Baltimore, MD, where it was confirmed that I had cystinosis. It became clear that an eventual kidney transplant would be necessary. This was very exciting because I knew the kidney transplant would change my life forever!

The goal of having a successful transplant was our primary objective in 1975. Since I was adopted, this could have made the process difficult, but it did not. Lynne Blissit, my biological mother was contacted and without hesitation offered to give me one of her kidneys. Fortunately, I only had to go through 3 weeks of dialysis before my transplant. At the time of transplant, at age 11, I was 3'11 and 39 lbs.

As an adult, I'm 5'3", 130 pounds and in excellent physical condition. As of this writing, I've had my transplanted kidney for 36+ years and it's working great!

After my transplant, I felt great except for my eyes. I was told that it was very unlikely anything could ever be done about my eye condition. I intuitively knew this was incorrect. For years, I had severe photophobia and eye pain that got worse with age. Many times it would feel like sand was in my eyes. Fortunately in June 1999, while doing an internet search, I discovered there was a cystinosis treatment available for my eyes at the National Institutes of Health (NIH). My first visit to the NIH was in September 1999. This was my first time to meet with Dr. Gahl and Dr. Kaiser.

Dr. Kaiser gave me Cysteamine eye drops, which instantly eliminated my pain and changed my life. One year later, my eyes were completely crystal free and have been crystal free ever since. Dr. Gahl put me on Cystagon® in 1999 in order to minimize future damage from cystinosis. Taking Cystagon® has been a challenge and I do the best that I can.

I graduated from the University of Texas at Arlington with a Bachelor of Science in Information Systems in August 1986 and have worked in this field for my entire career. I'm employed by Tarrant County College and I work for the Desktop Support Services department. I enjoy keeping up with my friends in the cystinosis community and offer advice based on my experiences. My personal interests include sports, working out, playing poker tournaments, and trying out new restaurants, and travel, with my favorite destinations being Vegas, Brazil, and Europe.

## FAMILY ISSUES

### Caring for Yourself

The parents of a child with cystinosis need to work through the stages of grief that may include denial, anger, bargaining, and depression. Parents may feel the need to grieve the loss of their “healthy” child and “idealized” normal life and rebuild a new “normal” around life with cystinosis. There is no time frame for this. As changes occur in the child's health, such as approaching transplant, the parent may revisit these stages of grief. <sup>[39]</sup>

Working through it all takes time, love and patience. It takes a good support network of family, friends and professionals. It takes an acceptance that whatever emotion one feels is okay. It takes the recognition that at times one needs to step back, regroup and recharge. It takes a willingness to ask for help and then be willing to accept it. There is truth in the statement: you can't take care of anyone else if you first don't take care of yourself. Keep a journal. Keep up with a hobby. Exercise. Meditate. Read or listen to audio books. Remember sometimes to just breathe.

### Faith and Hope

One of the hardest lessons for parents of children with chronic disease to learn is that, no matter how much they want to, they can't fix it or take it away from their child. They try, by giving medications, following treatment regimes strictly, and keeping doctors' appointments. They work hard to keep everything “normal” and do everything “just right”.

Sometimes, in spite of everyone's' best efforts, it is not enough. Stress gets overwhelming. Crises and losses happen. To watch your child suffering, and be unable to relieve that suffering, is at times unbearable. Many families rely on prayer, faith, and hope.

Your community of faith, and the cystinosis community itself, can provide vital support: comforting words, understanding, and prayers when you need them.

Whatever your spiritual beliefs, the challenges of living with this illness are likely to teach you the most profound lessons about life, love, courage, and community.

### Single Parents

If you are a single parent (or a military spouse whose partner is deployed!) with a child or children with cystinosis your support network needs to be a little bigger: family, friends, neighbors, employer, school, and perhaps respite services from your local county and state providers. Childcare is expensive so look for volunteers first! Reach out to those around you with a list of needs that you can entrust to someone else: shopping, school drop-off, making up meds, spending time with your child so you can take a shower or a nap, staying with your child while you run errands by yourself! The round-the-clock medication schedule is particularly challenging when you are the only one doing it. Lack of sleep can really take its toll. You need to trust others to help with the care of your child, and take care of yourself as well as you take care of your child. Also, reach out on message boards and Facebook – you'll find you aren't alone in going it alone.

## Caring for Your Marriage

Cystinosis affects the entire family, and results in major changes in schedules and priorities. Parents may spend a significant amount of time apart, one focusing on the child with cystinosis and the other handling the house, siblings, insurance issues and work. Both parents may be sleep deprived. Marriage issues can result. Marriages and relationships that were not stable prior to child's diagnosis of cystinosis may have great difficulty. Healthy marriages tend to become stronger. Parents who have remained in strong marriages for years while raising a child with cystinosis report that parents must take the time to recharge themselves, both individually and as a couple. Taking a break from the daily routine increases the patience, attention and positive energy they can offer to their children.<sup>[5]</sup>

### LIVING WITH CYSTINOSIS: LAURA'S FAMILY

Cystinosis has been tough on our family at times. During the first few years it was just coming to terms that this would be her life and insuring that she was always living the best life possible. I went through many stages, from super religious, to somewhat jaded, to somewhere in between. My husband was angry for many years and found his way to acceptance. Our marriage has ridden this journey fairly well. We have moments where we would like to strangle each other but we are in this fight as a family. We have made the choice to be married and fight to stay that way, as strongly as we have fought for Laura's well being. Sometimes it is not pretty, but we know our journey is better travelled together than separately.

Our son had a tough time during his teen years. He did not want to be the brother of the sick girl, and he made the choice to live with his father during this time. Our relationship was strained for many years. I can say now, at 23, Christopher loves Laura dearly. He organized and held a fundraiser for CRN with his platoon, without any input from me and without my knowledge until two days before the event! He spoke to his platoon about Laura and all those with cystinosis and told me that he found himself "choking up". He is very private about his feelings, so sharing with his platoon and becoming emotional was very powerful.

### LIVING WITH CYSTINOSIS: SARAH'S FAMILY

Cystinosis changed the whole dynamic of our family. For three years, my husband and I existed to keep Sarah alive. Our other children were amazing. They never complained about my time away from them or everything depending on Sarah. They quickly got used to Sarah vomiting in the car or at the store. In some strange way, it became our "normal". Our lives flowed around Sarah's needs. Our other daughters started doing school projects on cystinosis. Soon they were telling us interesting facts! Flexibility became our mantra because nothing ever went as planned. It still doesn't. With the very active children and one with special needs, my husband and I split duties and activities. We were rarely seen together because I was usually with Sarah. We laugh now, but one season, my son's entire baseball team and their parents thought my husband was a single dad! As important as flexibility is, humor is vital. After five years of living with cystinosis, we have settled into our own reality full of laughter, love, honesty, and maybe someday....a good nights sleep!

### LIVING WITH CYSTINOSIS: KACY'S FAMILY

At the time of diagnosis our sons were 10 and 12 years old. We tried to keep their lives as normal as they were before she was diagnosed. They both played travel sports and we continued with those. The medication routine was a crazy change, but they watched it all and it became normal for them. They also watched how Kacy just adapted to her new life. Although it was a drastic change from pre-diagnosis, she made it easy for us as a family. She went back to being the same spunky, high-energy girl she was before...just more high maintenance. Sometimes drastic changes can tear families apart, but in our case I felt like it brought my husband and I closer together and made our family unit tighter. We have all learned to be better, stronger, braver people from her.

## RESOURCES FROM CRN WEBSITE

The Cystinosis Research Network website is a wealth of information and resources for coping with the challenges of cystinosis. For more information, visit <http://www.cystinosis.org/> to check out available resources.

Article Library	Doctor	Medication	Stem Cells - NIH Info
Books on Tape	Recommendations	Management	Support Resources
Camps	Education	NIH Genetic Testing	Symptoms and
Cystinosis Brochures	Eye Drops	Registry Website	Treatment
CRN Fellow Contact	Financial	Cystinosis Information	Pediatric to Adult Care
Information	Flight Resources	Parenting and Families	Transitioning Guide
Cystinosis	Genetic Information	Pharmaceutical	Travel Tips
Organizations	Legal	Publications	Wish Organizations
Cystine Determination	Medicaid/Medicare	SSI	
Lab	Medical Home	State Facts for Health	
Disabilities	Medical Information	Insurance and Other	
Disability Law	Medical Search Sites	Health Info	



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302 Whytegate Court  
Lake Forest, Illinois 60045 USA  
Phone: 847-735-0471  
Toll Free: 866-276-3669  
Fax: 847-235-2773  
E-mail: [info@cystinosis.org](mailto:info@cystinosis.org)  
[www.cystinosis.org](http://www.cystinosis.org)



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