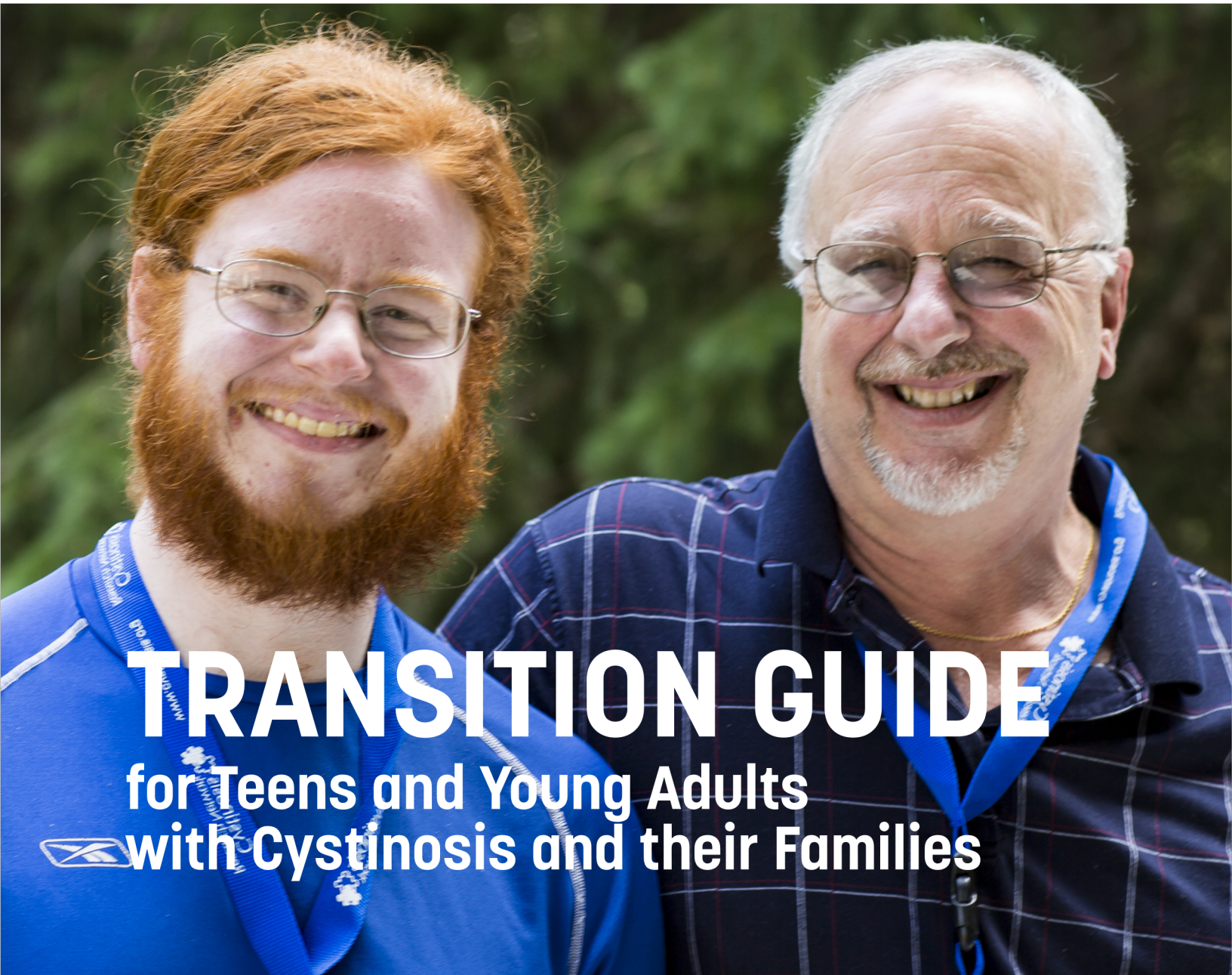
CYSTINOSIS
RESEARCH NETWORK

Dedicated to a Cure. Committed to Our Community.



TRANSITION GUIDE

for Teens and Young Adults with Cystinosis and their Families

CYSTINOSIS
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The Transition Guide was developed to assist teens, young adults, and their families learn more about cystinosis, empower them to take charge of their own lives and health care, and move successfully from pediatric to adult-oriented care.

The best transitions are PERSONALIZED and led by INDIVIDUALS with cystinosis and their FAMILIES. Teens and their parents can work as a team to reach the goals of healthy independence and management of their own health care.

We look forward to hearing from you, the pioneers on this journey. Please contact us with your ideas on how we can improve this Transition Guide.

Many thanks to all who contributed to this Transition Guide including individuals with cystinosis, their families, physicians, and the members of the Cystinosis Research Network Adult Care Excellence Initiative team.

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The Cystinosis Research Network's vision is the discovery of improved treatments and ultimately a cure for cystinosis. The Cystinosis Research Network (CRN) is a volunteer, nonprofit organization dedicated to advocating and providing financial support for research, providing family assistance, and educating the public and medical communities about cystinosis. We are a private, nonprofit 501(c) (3) corporation, Federal Tax ID 04-3323789.

The Transition Guide would not have been possible without the wisdom shared by the entire cystinosis community and the efforts of the members of the Cystinosis Adult Care Excellence Initiative. They are dedicated to advocating for individuals with cystinosis and their families. The members include Maya Doyle LCSW, Cheri Friend, Christy Greeley, Colleen Hammond RN, Shea Hammond, Jessica Jondle, MaryBeth Krummenacker, Pam Woodward, and Tahnie Woodward. A special thank you to William A. Gahl, MD, and Galina Nesterova, MD, for their contribution.

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THE JOURNEY TO ADULTHOOD

Becoming an independent adult is a long and winding road filled with changes and challenges for all of us. Having grown up with cystinosis, teens and young adults in our community may know more about their health, and about illness, than many of their peers. They may in many ways be wise beyond their years.

Parents work very hard to make sure their children have the resources needed to cope with cystinosis as they are growing up. It is likely that a family will develop strong, reciprocal connections with the medical specialists who have cared for a child for many years. Most medical institutions have policies concerning when young adults should begin their transition to adult-oriented primary care doctors and specialists; a patient may be 16, 18, 21, or 25 when the shift actually occurs.

What is Transitioning?

The term transitioning came into use in the late 1980s, inspired by Surgeon General C. Everett Koop, who observed that children whose lives he had saved through surgery as tiny infants were now surviving into childhood and adolescence. Koop described transitioning as the "one major issue" for chronically ill adolescents that had not been adequately addressed by the healthcare system[1]. In 2002, a consensus statement from pediatric and adolescent health professionals in the journal Pediatrics defined the goal of transitioning: "to maximize lifelong functioning and potential through the provision of high-quality, developmentally appropriate health care services that continue uninterrupted as the individual moves from adolescence to adulthood"[2].

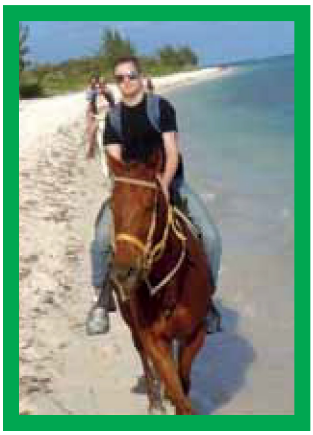
- The concept of transitioning may also include navigating a variety of educational systems, from secondary school or special education services to college or a vocational program.
- Some healthcare institutions have structured programs to help with transitioning, while others may require families to do more of the work. Either way, being knowledgeable about cystinosis, and learning to be your own advocate, are the keys to a successful transition to adult care.
- Each person and each family has a unique experience, based on individual medical and development needs, the support system in place, and personal style.

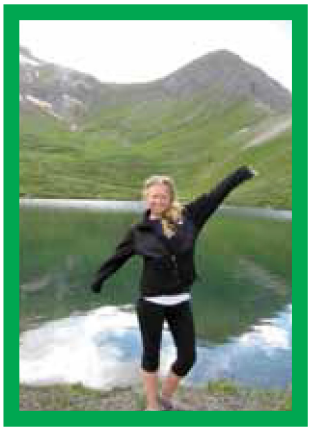
Transitioning – a Parent's Perspective

"The hardest thing I have ever had to do was trust that my son could manage his own healthcare needs – and then step back and let him do it. Sink or swim, his choices, his consequences… we fight all their lives to maximize the quality of their lives. We get them to 18 or 20 reasonably healthy and then have to step back as they are thrown into an adult system that is still learning about the disease they have, and expect them to manage something it has taken us decades to do. ***It requires a leap of faith as wide as the Grand Canyon"[3].***

"For parents, LETTING go is a process in which we change our relationship to our children and transfer responsibility for decisions concerning their lives from us to them"[4].

Transitioning – a Young Adult's Perspective





It's important to know where to go and what to do when you are grown and have to take care of yourself"[3].

"Having cystinosis has made me stronger and given me great coping skills, but at the same time it gets frustrating having to deal with all of the responsibilities and problems that come with it"[3].

Transitioning for Adolescents with Cystinosis – a Physicians' Perspective

From Dr. William A. Gahl and Dr. Galina Nesterova, National Institutes of Health

Dr. William A. Gahl

Cystinosis carries a significant burden for adolescents who require the attention of medical professionals throughout their lives. A major issue involves the transfer to adult medical care. In addition, young adults with cystinosis should thoroughly understand the disease and be able to describe it to others.

- Adolescents should learn about treatment options, know their medications and the side effects, familiarize themselves with the adult care unit and the consultants responsible, and know the procedures for making and keeping appointments.
- Another important competency involves maturely handling personal issues such as transportation to appointments and other logistic requirements related to the disease condition.
- Focus should be placed on the individual's positive characteristics, with an ultimate aim to build resilience, better compliance to treatment, and improved health outcomes.
- Many of the challenges associated with cystinosis may be minimized by having a supportive family and positive social peer support, and by engaging in activities that eliminate self-focus, such as volunteering in community activities.
- The young adult with cystinosis should also have sufficient knowledge about sexual health and the use of alcohol and drugs through discussions with a consultant.
- Primary physicians or endocrinologists knowledgeable in pubertal development and fertility should help care for both male and female adolescents.
- Cystinosis adolescents deserve empathy and respect for their disabilities and for their ability to cope with difficult situations. They should be engaged and encouraged to achieve successful management of their disease. Their positive assets should be nurtured to insure adherence and good outcomes.
- A planned, interdisciplinary approach to the care of these individuals should be designed with goals and objectives clearly delineated for all in a culturally and literacy appropriate format.

Nephropathic cystinosis is a chronic disease complicated by several issues, including its multisystem involvement, its requirement for both pediatric and adult medical care, and its treatment.

Dr. Galina Nesterova

Before the last quarter of the 20th century, cystinosis uniformly resulted in early mortality (early deaths), so that no patients required health care as adults. However, as a result of improved early diagnosis, successful renal (kidney) transplantation, and treatment with cystine-depleting agents, more children and adolescents with cystinosis are surviving into adulthood with improved lifestyles and a life expectancy beyond the fifth decade. For these individuals, the primary goal of medical care is to delay or reverse the deterioration of organs and tissues that results from intracellular cystine accumulation.

Several Factors Affect the Well-Being of Adolescents with Cystinosis.

These include:

- Medical/physical concerns
- Psychosocial functioning
- Long-term disabilities
- Multiple hospitalizations
- Treatment compliance
- Co-morbidities such as gastrointestinal complications with possible swallowing problems
- Hypertension (elevated blood pressure)
- Photophobia (lights hurting the eyes) due to corneal crystals
- Visual impairment due to retinopathy (involvement of the back of the eyes)
- Metabolic bone disease with skeletal deformities as consequences of rickets
- Persistent polyuria (large urine output) in pre- and post-transplant patients
- Endocrine involvement including diabetes and hypothyroidism (low thyroid function)
- Various degrees of myopathy (muscle involvement) affecting strength
- Delayed puberty (sexual maturation)
- Male hypogonadism (low male hormone levels) and infertility (inability to conceive)

These complications influence the choice of profession and the pursuit of family life, making adolescent maturation more challenging. Because of the rapid emotional, social, and psychological changes occurring at this time, together with pubertal changes in growth and strength, a number of problems may become unmasked during adolescence.

UNDERSTANDING CYSTINOSIS

It is important for children in this age group to have an understanding of cystinosis and how it affects their life. Having a disease that impacts multiple organ systems complicates this understanding.

How Do You Define 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 as follows:

Cystinosis is a rare 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. Without specific treatment, children with cystinosis develop end stage kidney failure at approximately age nine

In Their Own Words

Individuals with cystinosis have different ways of defining and explaining cystinosis to their families and friends. The following are some examples:

"She [an individual with cystinosis] just tells her friends that the filtering parts of her kidneys are broken and that hers don't work like theirs in filtering out the bad things and keeping the good. She shares that she has to take many meds in order to stay healthy."

"Mum would explain it as 'when you were born, your kidneys stopped working. You have this special condition called cystinosis – that is why you have to take medications – to help your kidneys to be healthy again.'"

"Basically, we all have something in our bodies (cystine) that healthy bodies know how to get rid of naturally, but mine doesn't. My body stores it as crystals, first in the kidneys and then in the eyes. Eventually that can lead to kidney failure and affect other organs. As long as I take my medication, it should be fine."

How Is Cystinosis Inherited?

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 and/or misdiagnosed. Scientists have mapped the cystinosis gene, CTNS, to chromosome 17p13.

Every person's body is made up of millions of tiny structures called cells. Each cell comes with a full set of instructions that 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.

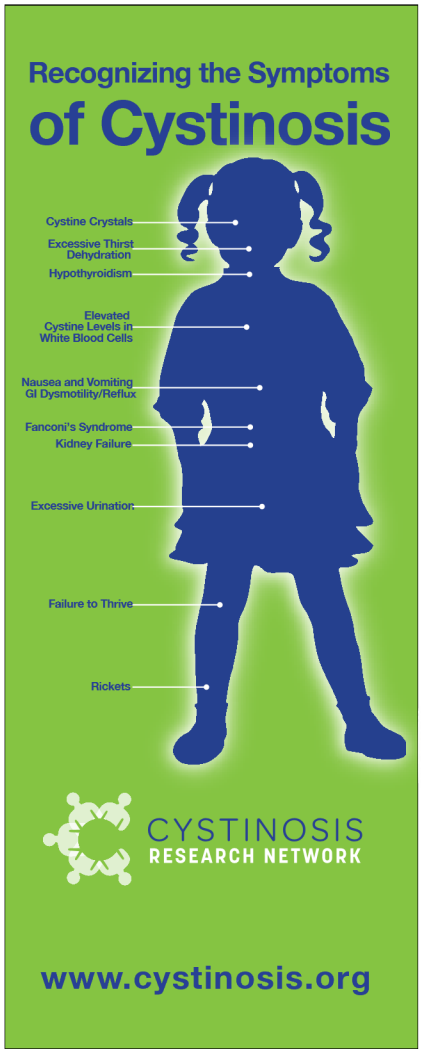
Genes are often called the units of heredity because the information they contain is passed from one generation to the next. We all get one gene in each pair from our mother and the other gene in the pair from our father. In this way, our bodies work with a combination of instructions inherited from 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 that may cause cystinosis in their children.

The genetic path of cystinosis is therefore impossible to predict, and having a child with cystinosis 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 and a 2-in-4 chance (50% risk) of having a child that is a carrier. Every healthy sibling of a child with cystinosis has a 2-in-3 chance (66% risk) of being a carrier, like his or her parents. Families who already have a child with cystinosis may choose to have prenatal or post-natal genetic testing to find out if a new baby is affected by cystinosis, so that treatment can be started quickly.

Cystinosis Symptoms



In addition to its impact on the kidneys, cystinosis can affect other organs of the body. Complications can include muscle wasting, difficulty swallowing, diabetes, vision problems, and hypothyroidism.

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 that others their age. They urinate frequently, have excessive thirst, and often seem fussy. At 12 months, they typically haven't walked and bear weight only gingerly. A major complication is renal tubular Fanconi Syndrome, a failure of the kidneys to reabsorb nutrients and minerals. The minerals are lost in the urine and must be replaced. Generally, children with cystinosis are picky eaters, crave salt, and grow very slowly. If left untreated, this form of the disease may lead to kidney failure by 10 years of age.

In people with ***intermediate or juvenile (adolescent)*** cystinosis, kidney and eye symptoms typically become apparent during the teenage years or in early adulthood.

In ***benign or adult cystinosis,*** cystine accumulates primarily in the cornea of the eyes, but other organs are generally unaffected.

Treatment of Cystinosis

Over the last 25 years, the prognosis of a child born with cystinosis has greatly improved. Cystagon, Procysbi and Cystaran, new treatments, and ongoing research efforts are giving hope for a bright future for children and adults with 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.

The course of the disease was dramatically improved by the discovery that cysteamine effectively removes cystine from lysosomes. This therapy alters the course of cystinosis, including delaying the development of kidney failure and other systemic manifestations. Currently, cysteamine is available in an immediate release form (Cystagon®) that requires dosing every six hours. A delayed release form of cysteamine bitartrate (Procysbi®) was recently approved and only requires dosing every 12 hours. Cysteamine therapy is associated with side effects, including gastrointestinal symptoms and a sulfur odor. Dosing of cysteamine is adjusted based on measurement of WBC cystine levels. Cysteamine therapy must continue after kidney transplantation since it is critical for ameliorating the widespread manifestations of cystinosis.

Cystinosis results in the accumulation of cysteamine crystals in the cornea, causing light sensitivity, pain and blurred vision. This is usually diagnosed by slit-lamp examination. Crystal deposition can be halted, and accumulated crystals dissolved, by frequent use of cysteamine eye drops (Cystaran®). Effective therapy is often achieved by dosing 8-10 times daily. Patients require regular visits with a general ophthalmologist or cornea specialist, typically on a yearly basis. The doctor will also check for optic nerve swelling, an uncommon, but important finding that requires referral to neuroophthalmologyor neurology for management.

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 (what's 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 several problems, including: 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 patient 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.

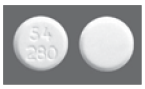
Although a new kidney will not be affected by Fanconi Syndrome or cystine accumulation, transplantation is not a cure for cystinosis and cystine will continue to build up in other parts of the body.

Kidney Transplant and Dialysis Resources

• American Association of Kidney Individuals	**http://www.aakp.org**
• American Kidney Fund®	**http://www.akfinc.org**
• American Society for Transplantation	**http://www.a-s-t.org/**
• Kidney School™	**http://www.kidneyschool.org**
• Kidney and Urology Foundation of America, Inc.	**http://www.kidneyurology.org**
• National Kidney Foundation®	**http://www.kidney.org**
• NIDDK Information Clearinghouse	**http://www.niddk.nih.gov/index.html**
• United Network for Organ Sharing	**http://www.transplantliving.org/**

COMMON MEDICATIONS FOR PEOPLE WITH CYSTINOSIS

Try **www.mymedschedule.com** to keep track of medications, doses, times, and purpose for each medication.

MEDICATION	APPEARANCE	PURPOSE
Carnitor® (Levocarnitine)	GENERIC available in liquid or tablet	Treats carnitine deficiency
Cellcept® (Mycophenolate mofetil)		Prevention rejection
Coenzyme Q10	GENERIC	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	GENERIC	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	GENERIC	May help to decrease urine output
K-Phos Neutral (potassium phosphate and sodium phosphate)		Electrolyte replacement – phosphorus.
Kaon-Cl (potassium chloride)	GENERIC	Electrolyte replacement – potassium
Lisinopril	GENERIC	Controls blood pressure
Magnesium	GENERIC	Restores low magnesium levels
Multivitamin	GENERIC	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	GENERIC	Promotes cardiac and joint health

MEDICATION	APPEARANCE	PURPOSE
Pepcid® (Famotidine)		Treats/prevents stomach ulcer/heartburn
Potassium Chloride		Electrolyte supplement
Prednisone		Prevents organ rejection
Procysbi® (Cysteamine Bitartrate)		Depletes cystine in the body
Prilosec® (Omeprazole)		Treats/prevents stomach ulcer/heartburn
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

EARLY ADOLESCENCE 12-15

Younger teenagers (ages 12-15) generally have some behaviors in common[6] , 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 oftentimes discouraging. Occasionally, medications are added to the already complicated medication regimen to offset the side effects of the treatments used to manage cystinosis.

Patients should learn tips for managing the various side effects of typical cystinosis medications :

- When recommended, take medications with food or just after a meal to reduce gastrointestinal issues
- Try to remain inactive for 30 minutes or so after taking medications
- 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[7].

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**[56]

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[8, 9]. There are alternate ways to teach almost everything[10]! 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.

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 nonadherence – try to understand what is happening, support rather than punish, 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.

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?

LATE ADOLESCENCE 15-18

Older teenagers (ages 15-18) generally have some behaviors in common[6], 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…?"
- Struggling to get conversation started? Try a game like The Ungame (several versions are available)

Taking (and Not Taking) Medications

For Teens

- Think about how much privacy is important to you at medication time. (See below.)
- 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.

MENTAL HEALTH

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[37], 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 elated 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 Cystagon® 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.

Conversation Starters and Exercises

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[12].

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. [38]

Emerging Adulthood

Emerging adulthood is a new term, coined by researcher Jeffrey Arnett, which is used to describe the experience of 18-30 year olds. These individuals are sometimes called the millenials, as they are entering adulthood at the beginning of a new millennium! What makes this age group unique, particularly in the U.S. and Europe, is that the years that were once thought of as a time of settling down, starting jobs, and building families is now often a time of extended exploration, trying out educational and career paths, traveling, forming relationships, and experiencing individual living situations[13]. These young adults may have expectations different from those of their parents or grandparents. Having grown up with cystinosis (and probably with a kidney transplant), a young adult may be eager to be more independent or may be feeling anxious about what the future will hold. He or she may also be facing some challenges that friends or healthy siblings will not experience.

Young adults may wrestle with:

- Negotiating what it means to be independent
- Allowing friends and family to provide help or support when needed
- Traveling, studying, moving away from home
- The changing roles of parent and adult child
- Making educational and work choices
- Starting relationships and families of their own
- Taking complete (or more complete) responsibility for their health and health care

Ups and Downs: Young Adults Speak Honestly About Cystinosis

- ***"The harder part has been getting myself emotionally and mentally healthy. In recent years, this has gotten easier too. The key to all of this has been my amazingly strong support network… It's been hard, but without all my experiences, I wouldn't be me[37].***
- ***"Generally I'm pretty happy with my life… but it's so much harder than anyone realizes[3].***
- ***"It [cystinosis] totally affects how I feel about myself – for the best. I am proud of all that I do in spite of cystinosis. It gives me a story of triumph and hope to share with others[9].***

Dating and Relationships

We asked adults with cystinosis for advice regarding dating and relationships, particularly when and how to tell the person you are dating about cystinosis. Here are their stories.

- ***"I've known my husband since I was 10. He used to come visit me while I was on dialysis in high school. So he knew from the start. When we split up and I dated other people in college, they always knew from the get-go. I think, at least for the past 12 years or so, I've always been very proud that cystinosis is a part of my life but doesn't dictate what I can and can't do. Therefore, I eagerly share with anyone who will listen. Until I was almost done with high school, though, I told no one about cystinosis. Of course, I ended up with someone who has been in my life for a very long time, which is very satisfying - because he had already seen me at my worst."***

- ***"Even if you don't tell the person right away, I think it's important to avoid deceit. The relationship cannot really become serious if you are withholding a part of yourself, and if the relationship becomes long-term and you are wearing some sort of mask, it will only hurt more in the end when the mask has to come off. For some people (like me), cystinosis is a very small part of their lives. But you have to realize that it might become a very large part of your life later,***

of what might be coming. My husband and I have discussed the general progression of cystinosis, the implications of having children and potential single fatherhood, the cost of health care, and many other related subjects.

- *"An important issue is the smell of Cystagon®. As I've talked with some of the young adults in our community, I've found more than one person who has stopped taking Cystagon® when embarking on a serious relationship. You cannot talk about dating without approaching the subject of close contact, kissing, etc. and Cystagon®. Quite frankly, if you're with someone who is going to make a stink (no pun intended!) about your lifeline, he or she isn't worth it."*

- *"I usually shared on the second or third date. My feeling is that if he doesn't stick around, then he is not the kind of person I would want to be with anyway. One time I waited too long, and the guy bolted shortly after I told him. Although he didn't admit it, I felt that was why. By then, I was more attached, so naturally it hurt more."*

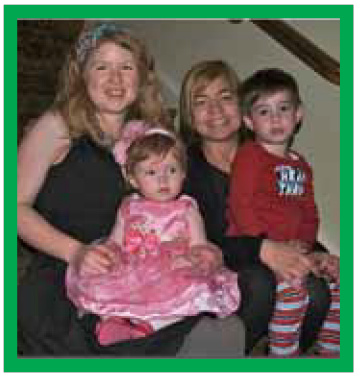
- *"Whether friend or boyfriend, I feel lucky that I have a reliable way to weed out the shallow people. When I tell others that I have cystinosis, and they are accepting of me, then I know they are open-minded people. Those are the kind of people I want as friends anyway."*

- *"I think you should wait until it feels right. I think it's better to let them get to you know you and what kind of person you are, and if there is chemistry, before you feel like you need to 'reveal it.'"*

Thinking About the Future

Reproductive Health and Cystinosis

By Dr. William A. Gahl and Dr. Galina Nesterova, National Institutes of Health



One example of a challenge faced by adolescents and young adults with cystinosis involves sexual maturity and the issue of reproduction. Untreated people with cystinosis nearly always exhibit late sexual maturation (puberty).

Males reach puberty at approximately 16-17 years of age, and may demonstrate a primary decrease in testosterone (hormone produced by testes) due to cystine accumulation in the testes. Poorly treated males exhibit high levels of luteinizing hormone and follicle stimulating hormone (other hormones needed for sexual maturation), as compensation for the low production of testosterone by the testes. Many patients benefit from testosterone supplements to restore secondary sexual characteristics such as facial hair growth, muscle mass, and increase in testicular (testes) size. Infertility is common in untreated males. Although the ability to have an erection (ability to have sexual life) apparently remains intact, no cystinosis patient is known to have fathered a child. Consideration should be given for sperm banking or the use of donor sperm for poorly treated male adolescents and young adults.

Girls with poorly treated cystinosis reach puberty at 14-15 years of age. Ovulatory cycles and gonadal endocrine parameters are normal in females, and several successful pregnancies and deliveries have occurred in women with cystinosis; the children have all been completely normal. Females should be informed that they have good chance to become mothers and that diligent cysteamine treatment will lessen the chance of cystinosis complications. There are no data on the teratogenic effects (risk of birth defects) of cysteamine in humans. Thus far, it has been recommended that women with cystinosis withhold cysteamine therapy when trying to conceive or during pregnancy.

FINDING ADULT SPECIALISTS AND EDUCATING PHYSICIANS ABOUT CYSTINOSIS

Choosing a New Primary Care Physician or Specialist

Whether you have insurance through your own employment or through your parent's plan, you will likely need to choose a Primary Care Physician (PCP). That person may coordinate your care and need to supply referrals, depending on your plan. The insurance company may be willing to have a nephrologist or other specialist coordinate your care, but you should try to find someone who will be a partner and advocate for you, regardless of prior knowledge of cystinosis. As part of a generation of adults living with cystinosis, you are also participating in educating a new generation of specialists and primary care doctors about the disease.

- Get the list of Primary Care and Specialty MDs from your company or insurance plan.
  - Most of these are also available online. Check with the insurance plan to make sure a physician you are choosing from the list is actively participating and accepting new patients!
- Find the physician's online bio, usually available within the website of their current academic appointment/medical center. This can tell you of any special interests and recent research.
  - Board Certification
- Internal Medicine is a more comprehensive certification than Family Practice
- Specialization – Look for an MD with additional certification in Nephrology or Endocrinology (or whatever specialty you feel would be most helpful for your unique situation)
  - Training
- Where did MD attend medical school and complete his/her residency?
- Any special interests, fellowships, or NIH training?
  - How many years has he/she been practicing medicine?
- Experienced
- New and willing to learn
- Has the doctor ever seen another cystinosis patient? Check with other cystinosis families in the area or visit the Recommended Specialists page at www.cystinosis.org
- Visit a website such as www.vitals.com and do a search for each person on the narrowed-down list of physicians that you are considering. This site has doctor ratings and feedback from patients. Look at reviews from people with complex medical needs.
- After you have selected a physician, fax or e-mail his/her office an introductory letter.
  - See Sample Introductory Letter on page 31 or at www.cystinosis.org.
  - Call and alert the office that the letter is coming. Ask to have the physician or office manager call you after it is received.
- Get a bad vibe on that phone call or after first visit? Keep looking and find the right "fit", as long as you have prescription refills and are not having a medical crisis!
- Have medical records or summaries from any and all specialists faxed to the new office prior to first appointment. Also, bring copies with you.

If you are female, you may need to choose between a Primary Care or OB/GYN doctor as your Primary Care Physician. A Primary Care doctor may be better able to coordinate specialists, but you also should have an OB/GYN, whether or not you are pursuing reproductive options.

What to Take with You to a Medical Appointment with a New Doctor

- Information about medical history, medications, and contact information for all physicians, insurance, and pharmacies. See the One-Page Health Information Sheet on the next page (also available at **www.cystinosis.org**
- A copy of records or a detailed summary from previous primary care doctors and specialist(s)
- Copy of insurance cards, including Medicare, Medicaid, and drug plans (if applicable)
- Cystinosis Overview Article – available at **http://www.cystinosis.org/**
- ***2002 New England Journal of Medicine: Cystinosis*** Dr. William Gahl, Dr. Jess Thoene, and Dr. Jerry Schneider[5]
- Articles related to any specific concerns you have – see article library at **http://www.cystinosis.org/**
- A notebook and a pen! Bring questions and take notes about the doctor's answers and any changes to your regimen.

What to Expect

- Expect your pediatrician or pediatric specialist to make contact and share information with new doctors before your visit. Ask for a shared visit with both providers if it is feasible. Some medical centers will automatically do this as part of your transition plan.
- Adult healthcare settings will generally be busier, and visits may be shorter. Expectations are high that someone who has grown up with a chronic condition will be knowledgeable about that condition and about taking care of himself or herself.
- While it is perfectly acceptable to bring a friend or family member along to a visit, healthcare providers will expect the patient to be able to answer – and ask – questions themselves.
- If you want your new physician or team to share information with your parent(s), you may need to sign a release or document that this is acceptable to you.

Sample One-Page Health Information Sheet

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

Sample introductory Letter

(Date)
Dear Dr. _____,

I would like to introduce myself as a new patient. As I reviewed the *(PCP/ specialist)* listings within my insurance plan, you caught my eye as someone that I would entrust with my care. *(or "you were recommended to me by _____").* Because my medical situation is complex, I would like to share some preliminary information with you prior to my first appointment on _____.

I have a condition called nephropathic cystinosis, a rare metabolic disorder that creates a buildup of the amino acid cystine in the body's cells. Since my diagnosis at the age of _____, this disease has been the focus of my medical care. What makes cystinosis a challenge is that it impacts several major organ systems.

For more information about cystinosis, the following excellent resources are available:

- **http://www.cystinosis.org (a list of research based articles and links to organizations involved with cystinosis research and support**
- **Numerous articles can be found using the search term "cystinosis" on http://www.nejm.org and http://www.springer.com**

Because everyone who has cystinosis is an individual, I also want to share with you some of my personal medical history. I started cysteamine therapy (the only approved treatment for cystinosis patients) at _____ and went into ESRD when I was _____. I received a kidney transplant in _____. Since that time, _____

My current health concerns revolve primarily around other cystinosis-related symptoms, including _____

I typically receive referrals to specialists in

_____

_____

(Examples: nephrology, endocrinology, ENT, ophthalmology, cardiology. You can also include" my current primary care doctor/specialists are:" and their contact information)

I have attached a list of my current medications for your perusal. Some of these medications must be acquired through specialty and/or mail-in pharmacies, and I am also providing their contact information.

I am knowledgeable about my medical condition and I strive to be pro-active and pre-emptive with my medical care. I look forward to my appointment with you.
Sincerely,

(name)

(contact info)

Educational Settings – College, Trade Schools, Vocational Programs

The Rehabilitation Act of 1973 (Section 504) and the Americans with Disabilities Act of 1990 (Title II) prohibit discrimination based on disability. "Disability" can include "invisible" conditions like cystinosis, and includes more "obvious" disabilities such as vision problems, learning disabilities, difficulty walking, etc.

Colleges and universities must have an Office for Students with Disabilities (actual names will vary). Consider documenting your health condition with your educational institution. This is important if you think you will need any "accommodations" to complete your studies, or if you think your attendance may be affected by illness. If you do not document your health condition ahead of time, your professors and administration will not be required to make special exceptions.

Under new federal regulations, insurance plans offered by colleges and universities will have to offer the same coverage as commercial plans – such as not imposing limits on lifetime costs or pre-existing conditions.

You can find out more about the rights of students with disabilities at **http://www.ed.gov/ocr/transition.html**

Cystinosis, Work, and Insurance Coverage

Individuals with cystinosis in the workforce may experience a variety of unique issues and concerns. However, career choices should be based on each individual's intellect, ability, interests, and life goals. People with cystinosis are not limited in choice of employment and work in a variety of professional and technical jobs.

The Interview

Aside from preparing for interview questions, showing up well-rested and ready to interact with a potential employer, and dressing to impress, young adults with cystinosis should know the state and federal laws related to questions that can be legally asked (**www.ada.gov or www.wwoc.gov**). Should information about cystinosis be disclosed at the interview? Probably not. Sharing this information at the interview can put both the interviewee and the company in a difficult position. The decision to hire should be based primarily on an individual's ability to perform the tasks of the job.

Choosing Health Insurance

Once a job offer is made, there are important health insurance issues to consider. First and foremost, an individual with cystinosis will need to know if there is employer-provided insurance. If so:

- Does the plan cover your prescriptions?
- Are your medications on the "formulary" or must they be ordered through a specialty pharmacy? Will you have to pay out-of-pocket for specialty pharmacy orders?
- Is there a mail-order option? Cystagon® and cysteamine eye drops will likely require those extra steps.
- Does the plan permit you to see the providers you already have established relationships with and allow you to be hospitalized at the medical center of your choice?
- Does the plan require referrals for each specialist visit, or do you simply need to choose "in-network" specialists?
- What does it cost to go "out-of-network"?
- For medications that may not be FDA approved, what will be the cost to you?

Remember that this process can apply to people of all ages who find themselves with a new job and/or a new insurance plan that require a change of medical provider or pharmacy.

Legal Protection

There are many laws that assist people with cystinosis in the workplace and with health insurance. As you begin your career, it is important for you to know these laws and how they impact you. A good resource about issues and legislation affecting young adults is **www.younginvincibles.org.**

The following is a brief description of these laws:

- **Section 2714 of the Affordable Care Act of 2010** requires all plans offering dependent coverage to allow individuals up to age 26 to remain on their parents' health insurance.
- **Section 2301 of the Reconciliation Act of 2010** includes a requirement for existing health insurance plans to provide coverage for adult children up to age 26 and eliminates the requirement that adult children be unmarried. The extension of dependent coverage provisions also means that adult children do not have to be in college full-time to maintain coverage under their parents' coverage. A good resource for information about healthcare reform is the Commonwealth Fund (**www.commonwealthfund.org**)
- **Consolidated Omnibus Budget Reconciliation Act (COBRA) of 1986.** This law allows you to continue your insurance coverage for up to 18 months through your employer if your job is terminated or your hours are reduced (**www.cobrainsurance.com**)
- **American with Disabilities Act (ADA) of 1990.** ADA requires that employers provide reasonable accommodations for individuals with disabilities who can perform the essential functions of the job (**www.ada.gov**).
- **Family and Medical Leave Act (FMLA).** This act permits employees to take up to 12 weeks of unpaid leave during a 12-month work period for an illness or to care for a spouse or child with an illness (**www.dol.gov**).
- **Ticket to Work and Work Incentives Improvement Act**. If you receive Social Security Income or Social Security Disability Benefits, this law may allow you to work and continue some of your insurance benefits. It also has a voucher system, where you can gain rehabilitation and other services to assist you in getting a job (**www.ssa.gov**).

Other Legal Issues

Health Care Proxy and Living Will

When you visit a new doctor's office, or are hospitalized, you may be asked if you have a Healthcare Proxy or Living Will. Everyone over 18 should think about who would make medical decisions if he or she is unable to (due to being in a coma, for example), and if there are certain treatments that the patient specifically does or does not want.

A Healthcare Proxy is a legal document that lets you name a healthcare agent – that is, someone you trust to make healthcare decisions for you if you are unable to make decisions for yourself in the future. It also gives you the option of listing specific healthcare wishes.

A living will allows you to leave written instructions that explain your healthcare wishes, especially about end-of-life care, should you be unable to speak for yourself in the future.

Visit **www.doyourproxy.org** to draft your own healthcare proxy or living will, or speak to the social worker on your healthcare team.

Guardianship

What is Guardianship?

All persons have a right to make decisions. If a person cannot make decisions because of mental incapacity or a developmental disability, the law can take away or "limit" a person's right to make decisions, and appoint someone else to make decisions. This is called guardianship[14].

There are several types of guardianship. Guardianship of the Property applies to decisions related to financial matters. Guardianship of the Person applies to decisions related to personal life, such as where one lives, what health care is provided, and where one works or goes to school. Limited guardianship applies to some, but not all, decisions related to personal life. Healthcare guardianship is an example of a limited guardianship. If healthcare guardianship has not been established, parents cannot legally communicate with healthcare providers about their adult child without the written permission of the young adult patient. This is because the Health Information Portability and Accountability Act (HIPAA) states that at age 18 years, youths who are capable of understanding the HIPAA privacy form must give written consent in order for records to be shared with others.

EXPLORE THE WORLD

Traveling

Having cystinosis should not prevent you from traveling and seeing the world. However, there are important steps you should take to ensure a safe, enjoyable traveling experience.

- Always include several days' worth of medication in your carry-on luggage. Even if you are taking a very short flight, there is always the possibility that checked baggage can be lost, and in certain rare situations you may find yourself without your items for a few days or more.
- If you are traveling with liquid medications (such as cysteamine eye drops), you should be aware that even with the current carry-on item restrictions, it is still your right to board a plane with these drugs. Make sure the bottles are clearly labeled with the prescription information, and carry written orders from your physician in the unlikely event that you must provide evidence of your need to be in possession of these liquids.
- Carry an emergency information card in your wallet or purse. If you are immunosuppressed due to transplant medications, make sure that information appears prominently on the card. Drug and food allergies should also be included. Think about what you would want an emergency medical team to know in the event that you are unable to communicate.
- If traveling abroad, check with your health insurance provider to see if you have coverage in your destination country. You may also want to look into traveler's insurance.
- Make sure you are up-to-date on any immunizations you may need for traveling internationally. You can "find health precautions and immunization recommendations for your destination country at **http://wwwnc.cdc.gov/travel/**
- Remember that in many countries, tap water may not be an option and in order to stay hydrated, you may need to purchase bottled water.

CLOSING THOUGHTS

Being knowledgeable about cystinosis, having an awareness of your own unique health needs, and planning ahead will allow you to manage your disease and meet the goals you set for yourself. By taking a leading role in your transition from pediatric to adult care, you will be empowered to maintain a full and independent lifestyle, without compromising your health. Remember that the transition from adolescence to adulthood is one that everyone must undertake, and cystinosis, while presenting a particular set of challenges, does not have to be a burden. Ultimately, following the suggestions in this guide and proactively serving as your own advocate will keep you on the pathway to success.

Questions, comments, or thoughts about how to improve this guide? Please email **transitioning@cystinosis.org**

TRANSITIONING PROGRAMS AND RESOURCES

Transition Literature on PubMed:
http://www.ncbi.nlm.nih.gov/sites/entrez?db=pubmed&cmd=historysearch&querykey=1

Health Care Transition Initiative at the University of Florida
http://hctransitions.ichp.ufl.edu

From Children's Hospital of Philadelphia:
http://www.chop.edu/service/transition-to-adulthood/resources-for-patients-and-families.html

Adult Health Transition Project, University of Washington
http://depts.washington.edu/healthtr/

Medical Home Portal
http://www.medicalhomeportal.org/living-with-chil

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