

# Living with Cystinosis

Survey Results  
January 2011

Cystinosis  
Research Network 

*Dedicated to a Cure. Committed to our Community.*

# Cystinosis Adult Care Excellence Initiative

February 2011

Due to the tremendous medical advances being made in the cystinosis community, our children are surviving well beyond adolescence. In the past, we have focused our discussions and hopes on our children's survival into adulthood. Our goal now is that they *thrive* into young adulthood and beyond. The cystinosis community must make key investments in adult care programs to meet the shifting needs of the adult population. The Cystinosis Research Network (CRN) has formed a working group to address the myriad challenges and tasks associated with the transition to adulthood, in order to provide our children with the tools required for this change.

The Cystinosis Adult Care Excellence Initiative was established to address at least the following three issues:

## **1. Transition from Pediatric Care to Adult Care**

Infantile Nephropathic Cystinosis was once thought of as a condition exclusively affecting children. Advances in treatments have allowed more and more persons with cystinosis to live well into adulthood. This presents individuals with cystinosis many opportunities and challenges, including how to incorporate treatment into an increasingly independent way of life. The transition from pediatric care to adult care can be difficult for young adult patients. Adolescents are accustomed to their parents communicating with the physician and managing their day-to-day health care. As patients approach their 20s, that responsibility shifts from the parent to the patient. It's a big change that can be scary for the patients and their parents. The transition requires open communication and education for young adult patients. The CRN is creating a Transitioning Guide to assist in this process.

## **2. Clinical Practice Guidelines for Cystinosis – Pediatrics through Adulthood**

The Clinical Practice Guide is to be used as a resource for adults with cystinosis, their parents, and their medical caregivers. Patients with cystinosis have, until recently, spent their lives under the care of pediatric specialists. Now that patients are living into their 40s and beyond, Internists are more likely to see adults with cystinosis.

### **3. Identification of Research and Treatment Needs to Improve the Quality of Life**

To determine the research and treatment needs of the adult cystinosis community, a detailed investigation of the effects of cystinosis on each organ system and the psychosocial effects should be developed. Investigation should be made to determine if treatments are available for the complications and if no treatments are available, request for research proposals should be initiated when financially possible.

To begin the identification of research and treatment needs to improve quality of life for adults with cystinosis, The Living with Cystinosis Survey was conducted in November and December of 2010. Nearly 150 responses were received. The survey results help strengthen our understanding of issues of concern to the cystinosis community and how cystinosis affects the lives of patients of all ages and their families. The information collected from this survey will be used to design and support programs, services, patient education materials, and advocacy efforts on behalf of individuals with cystinosis.

We are pleased to present to you the results of The Living with Cystinosis Survey. We hope the information collected here will move us closer to accomplishing not only our goals of improving quality of life for adults with cystinosis, but also our greater vision and mission.

**Christy Greeley**

**President and Executive Director, CRN**

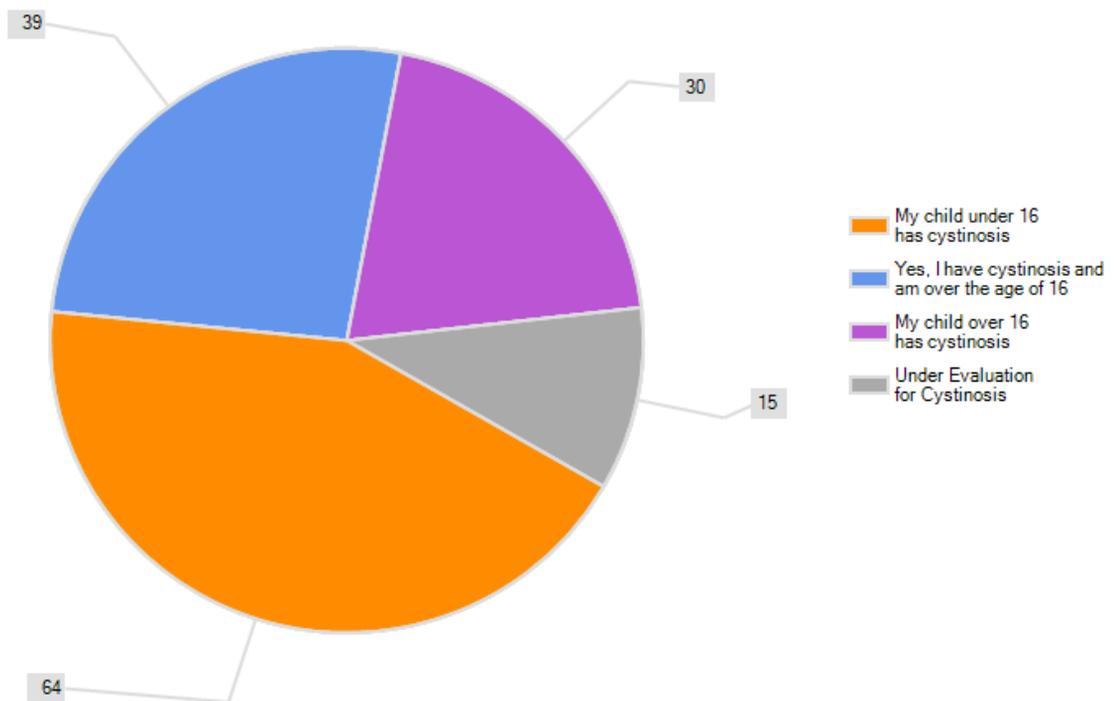
The Cystinosis Research Network is a volunteer, non-profit organization dedicated to supporting and advocating research, providing family assistance, and educating the public and medical communities about cystinosis. The CRN's vision is the discovery of improved treatments and ultimately a cure for cystinosis.

# The Living with Cystinosis Survey

- The Living with Cystinosis Survey was conducted online in November and December of 2010.
- This report will address areas of concern for several groups:
  - Children and adolescents, ages 0-5, 5-12, and 12-15
  - Older adolescents, ages 16-18
  - Emerging adults, ages 19-25
  - Adults, ages 26-40+
- The “voice” of patients and families affected by cystinosis is heard through selected quotes taken from answers to open-ended questions within the survey.
- The Living with Cystinosis survey was created by the transitioning workgroup of the Cystinosis Adult Care Excellence Initiative, with assistance from our professional advisor, Maya Doyle, LCSW and our scientific advisors, Ewa Elenberg, MD and Rick Kaskel, MD.

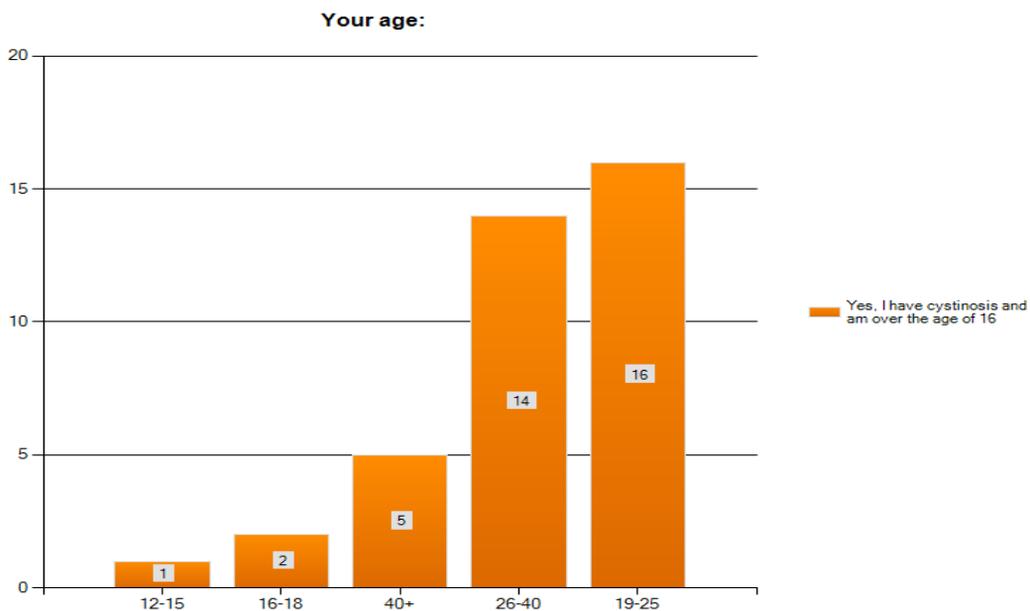
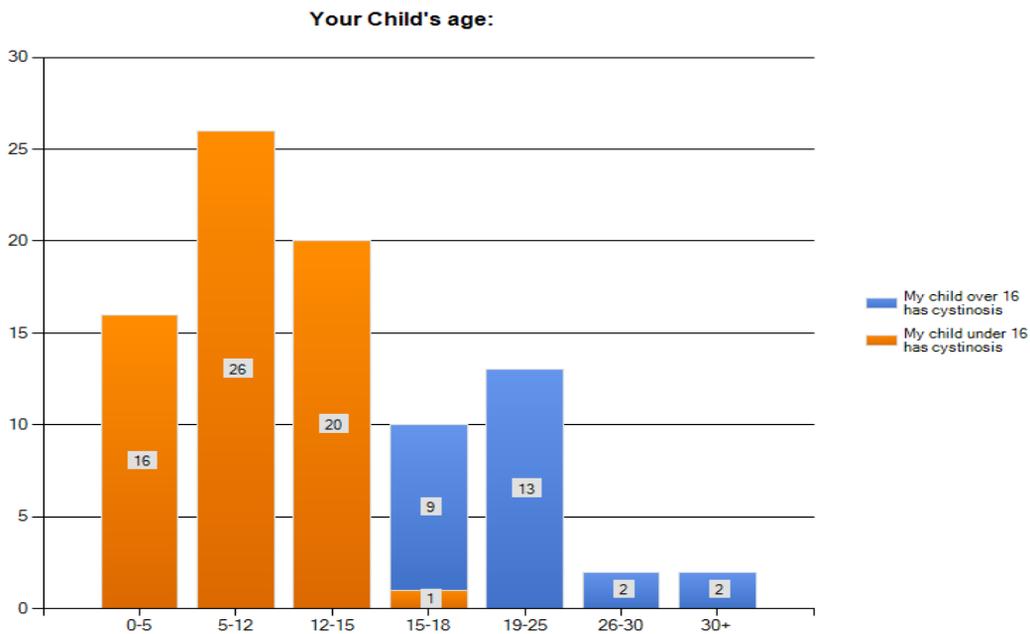
# Survey Responses

Have you been diagnosed with cystinosis?



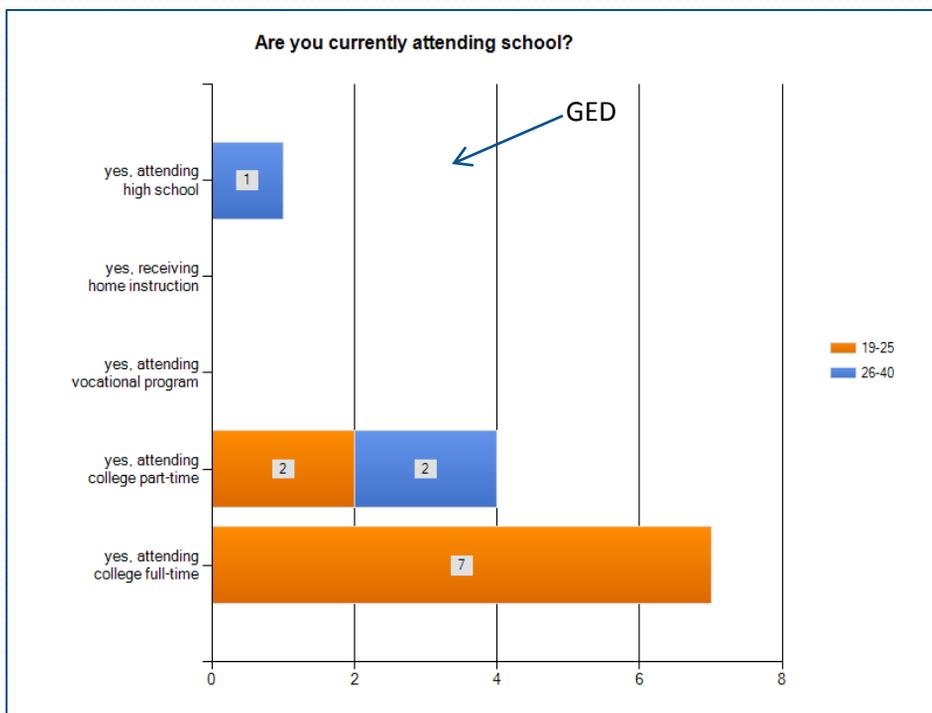
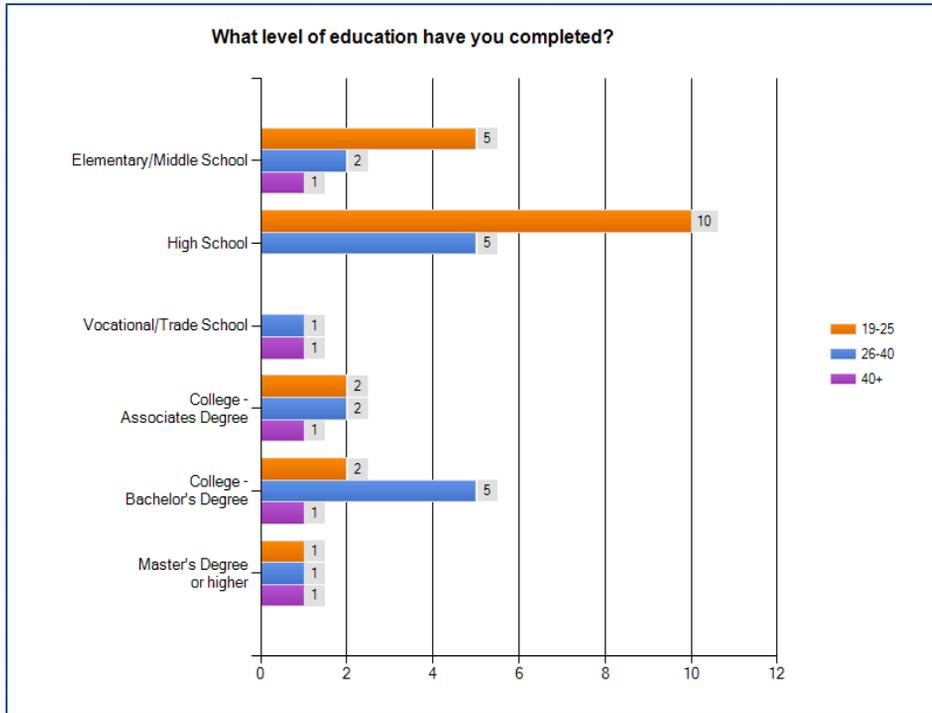
The Living with Cystinosis Survey received nearly 150 responses. While most responses were from the United States and Canada, we also received responses from Australia, New Zealand, Brazil, Norway, Denmark, Germany, France, South Africa, and the United Kingdom. Respondents were divided into three groups: those with cystinosis, parents with children ages 16 and under, and parents with children older than 16. Those currently under evaluation for cystinosis did not complete the survey but were referred back to the CRN website for further information.

# Respondents



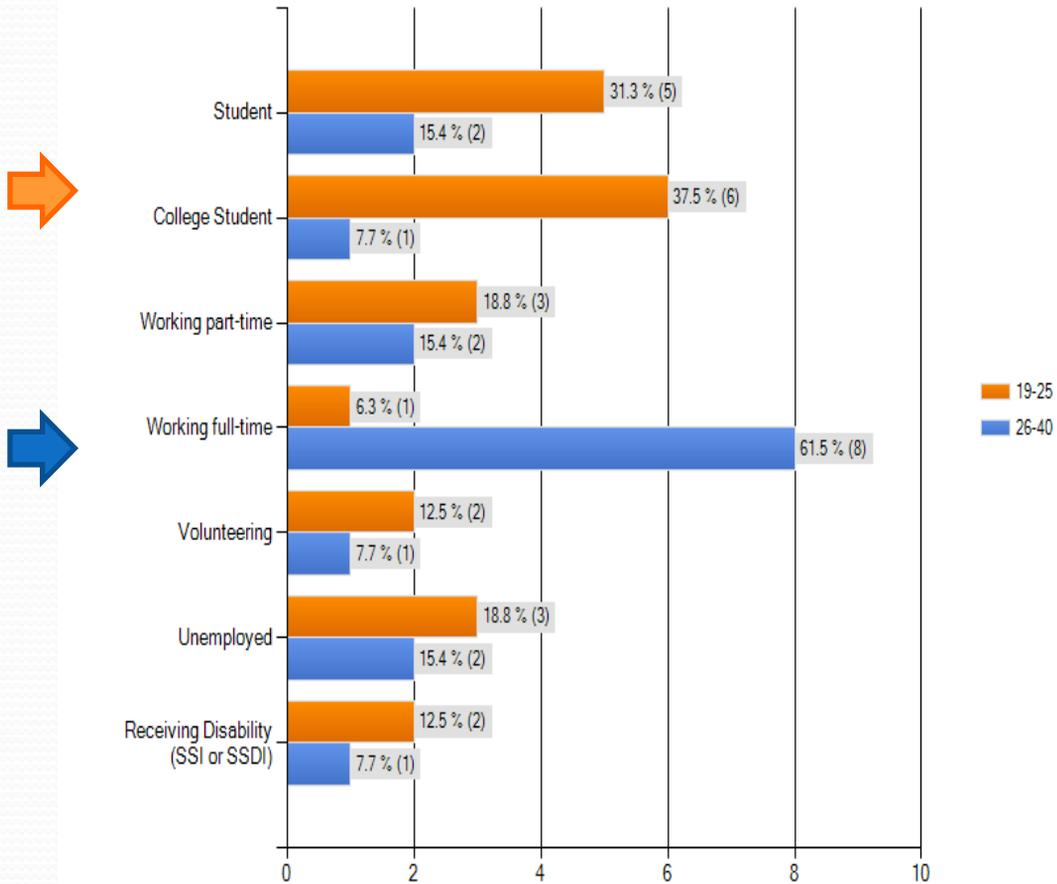
Respondents included parents of individuals with cystinosis below and above the age of 16, as well as individuals with cystinosis ranging in age from 16 to greater than 40.

# Educational status (adults/emerging adults)



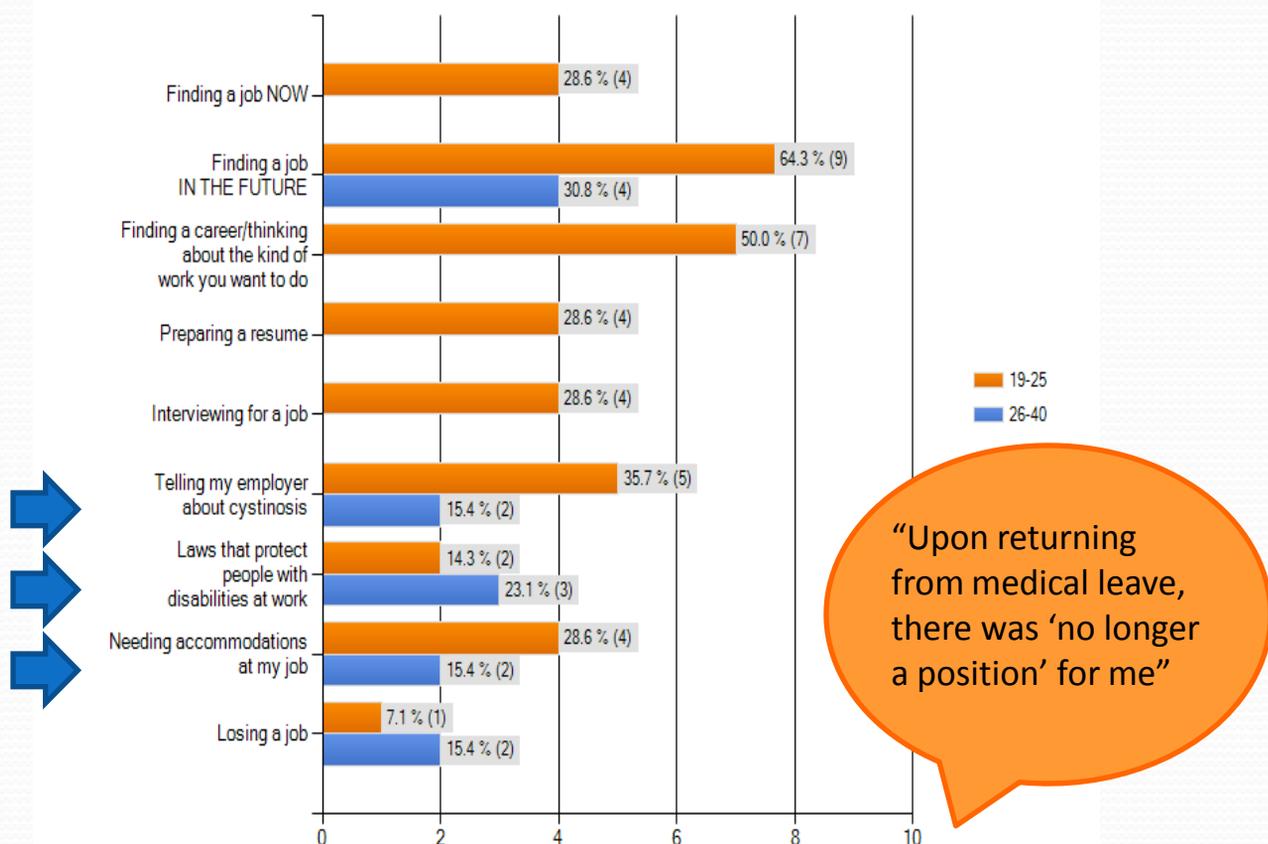
# Employment status (adults/emerging adults)

Current level of employment:



# Employment concerns (adults/emerging adults)

Current concerns about WORK and EMPLOYMENT

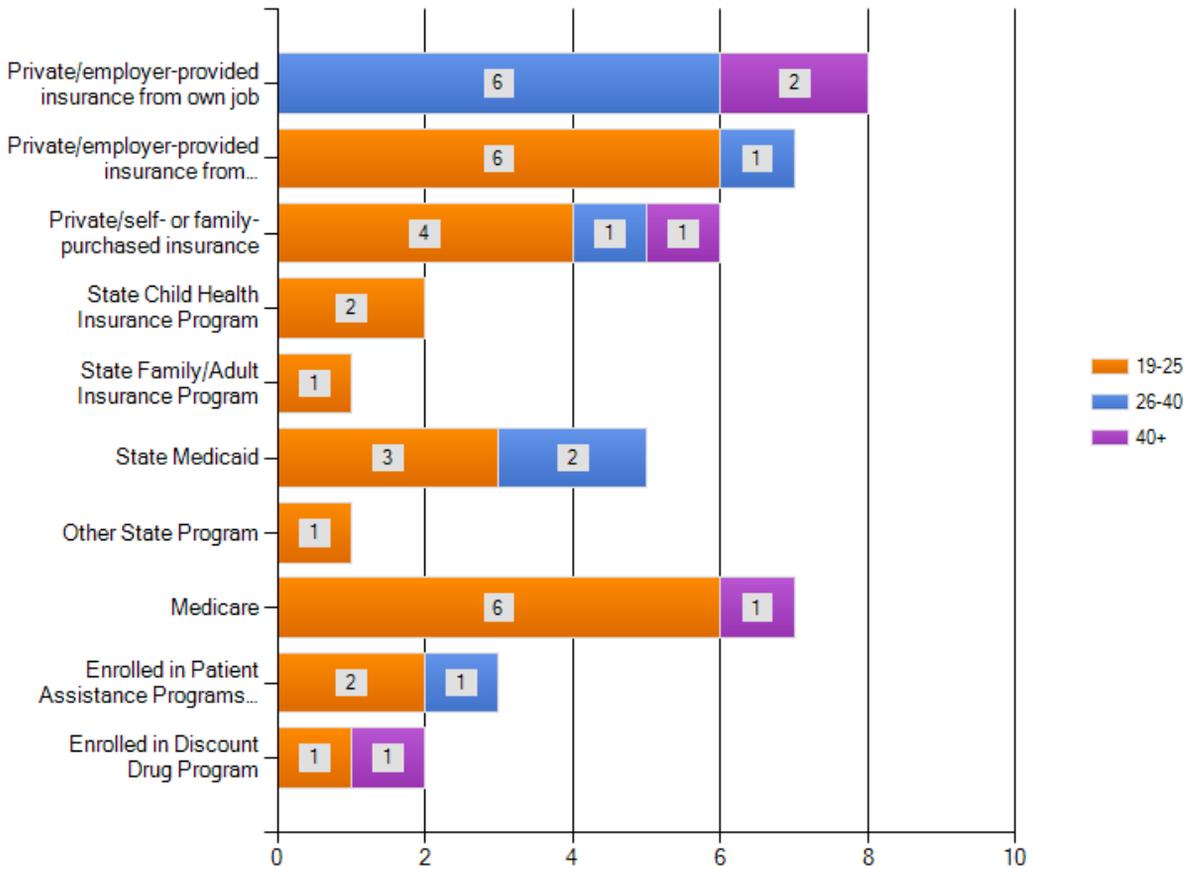


Many of these concerns – such as finding a job in the future, choosing a career, or preparing a resume – are developmentally appropriate tasks for *emerging adults*. However, the challenge of balancing work with health and health care needs, and of addressing discrimination and needs for accommodation at work, requires planning and knowledge of laws and resources.

"Discrimination... unable to advance because of health problems. How others treat me at work because of my health or missing work... some are very nice, some are terrible. I never knew people could lack so much compassion."

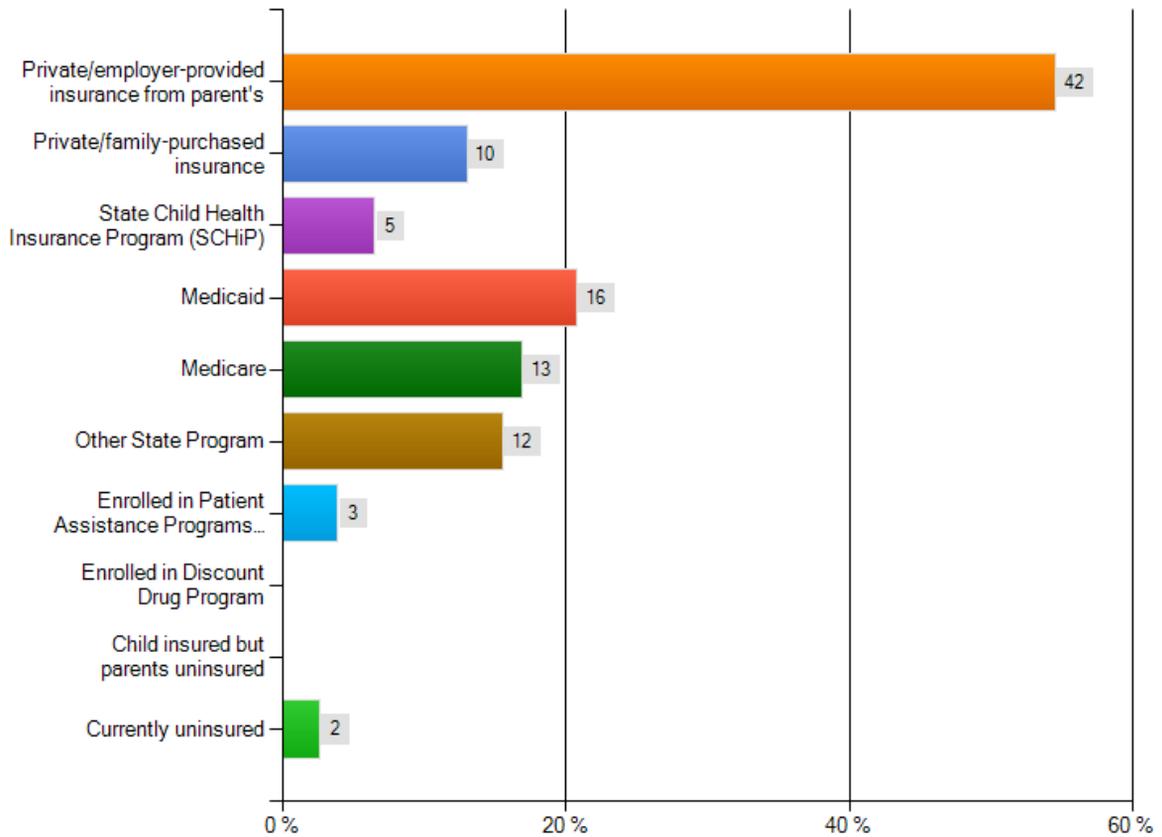
# Insurance status (adults/emerging adults)

Current insurance status (select all that apply)



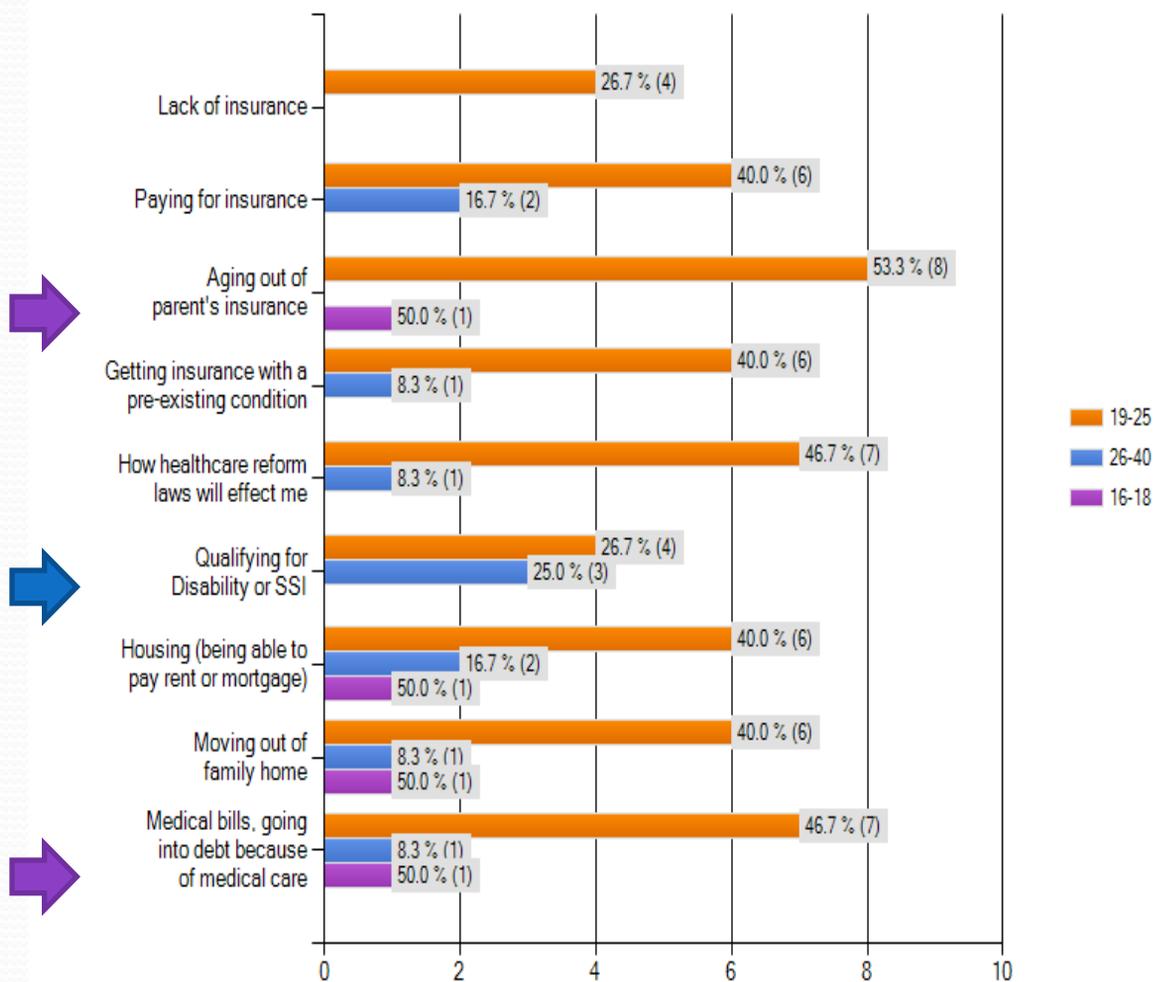
# Insurance status (children under 18)

Your child's current insurance status (select all that apply)



# Financial concerns (adults/emerging adults)

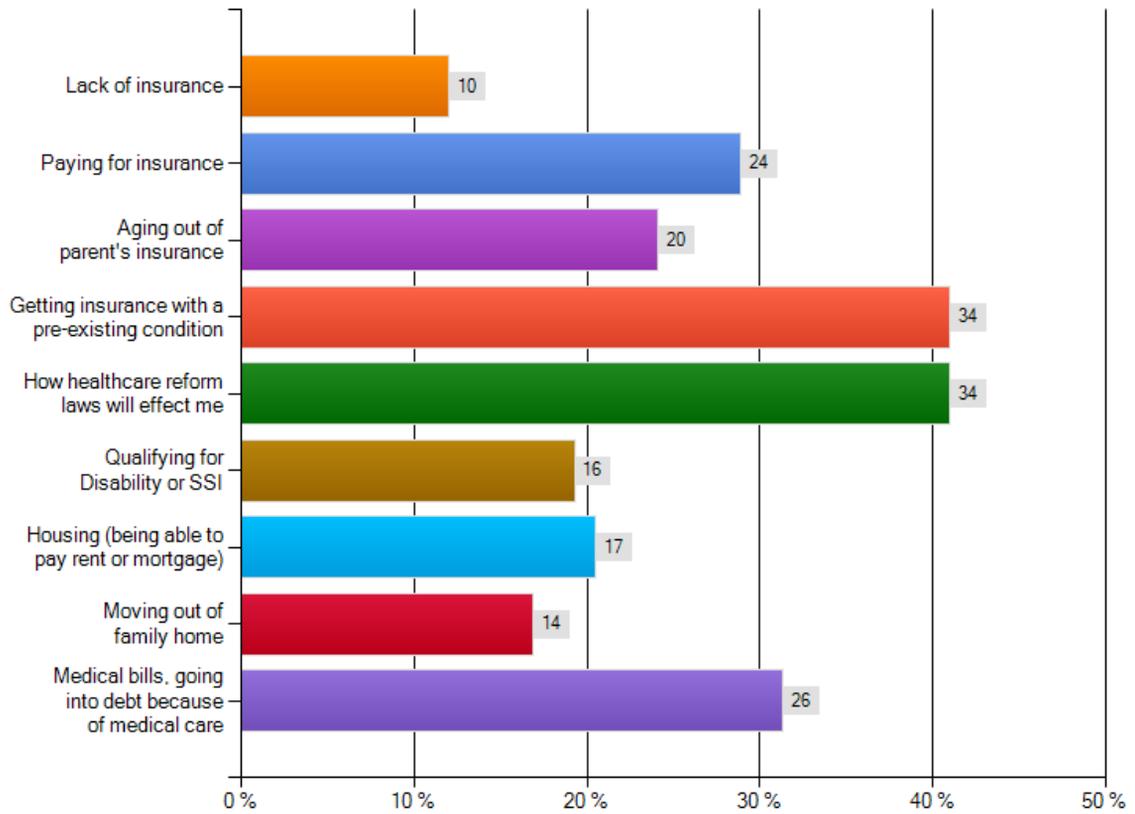
Current FINANCIAL concerns



Emerging adults with cystinosis are particularly concerned about financial issues and insurance. Their ability to move towards independence may be hampered by their ability to obtain health care and medications. Changes to dependent coverage options following health care reform will help some patients, but not all. Gaining information about laws and resources is vital, as is being advocates at the individual AND the policy level.

# Financial concerns (families)

Current FINANCIAL concerns



## Finances and insurance: In their own words

“...It's a little unnerving knowing that having a medical illness from birth can keep us from getting access to the meds we need to survive. The thought that once I'm thrown off my parents' insurance when I'm 26 I'll have to find a job/win the lottery to *get insurance or die* is kind of hard to take.”

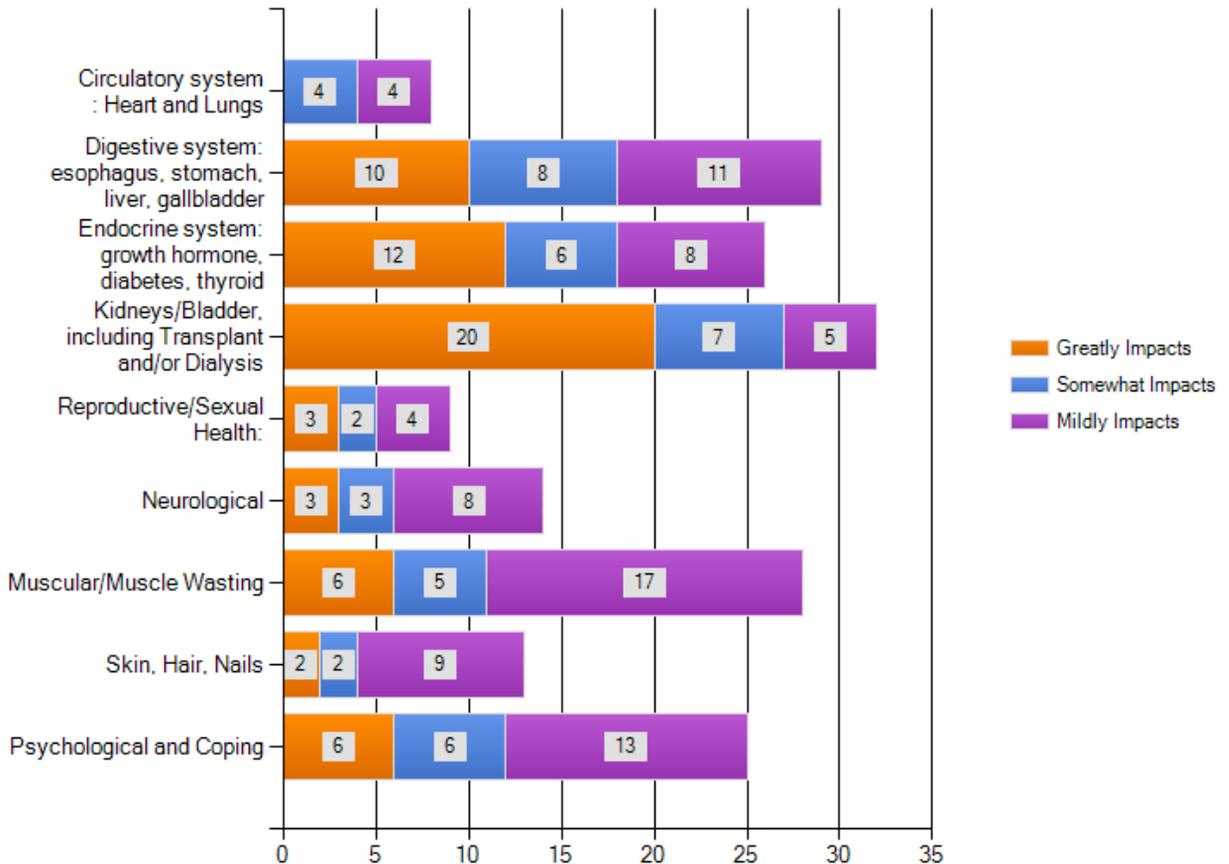
“[Resolving the issue of] health insurance [is critical]... so the transition from dependent life to independent is not so *terrifying*.”

“Remove the fear of losing insurance when he no longer is eligible to be covered under our policy. With the current economic situation, relying on employer-based policies is *terrifying*.”

“If [they] didn't need to worry so much about what's going to happen financially, they could focus on [their] health [and] his new transplant. They are both working, both have insurance, he has Medicare, and yet [he is being told] that he can't have the transplant he needs right now because of Medicare issues... *That could mean dialysis or death. It is a national disgrace*.”

# Complications (children)

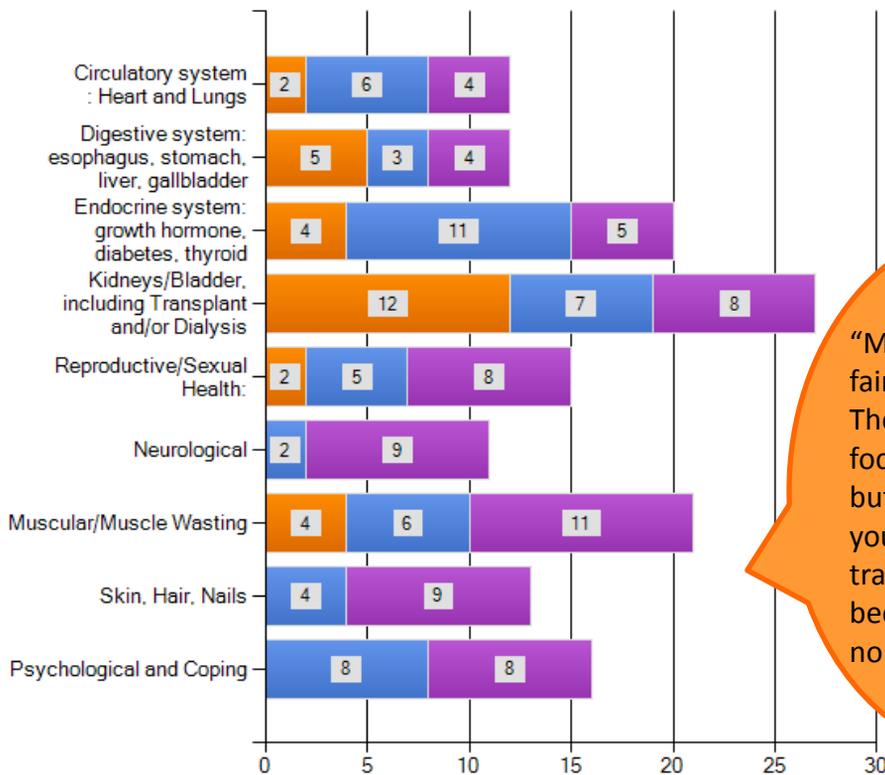
To what degree do complications affect your child? Complications can be from illness or from medications.



For families with children under 18, loss of kidney function (and the need for dialysis and transplant), growth and nutrition, and digestive problems were the greatest concerns. Vomiting and issues related to gastrostomy tube (placement, dependency, impact on activity) was a particular concern for parents of younger children.

# Complications (adults)

To what degree do complications effect you? Complications can be from illness or from medications.

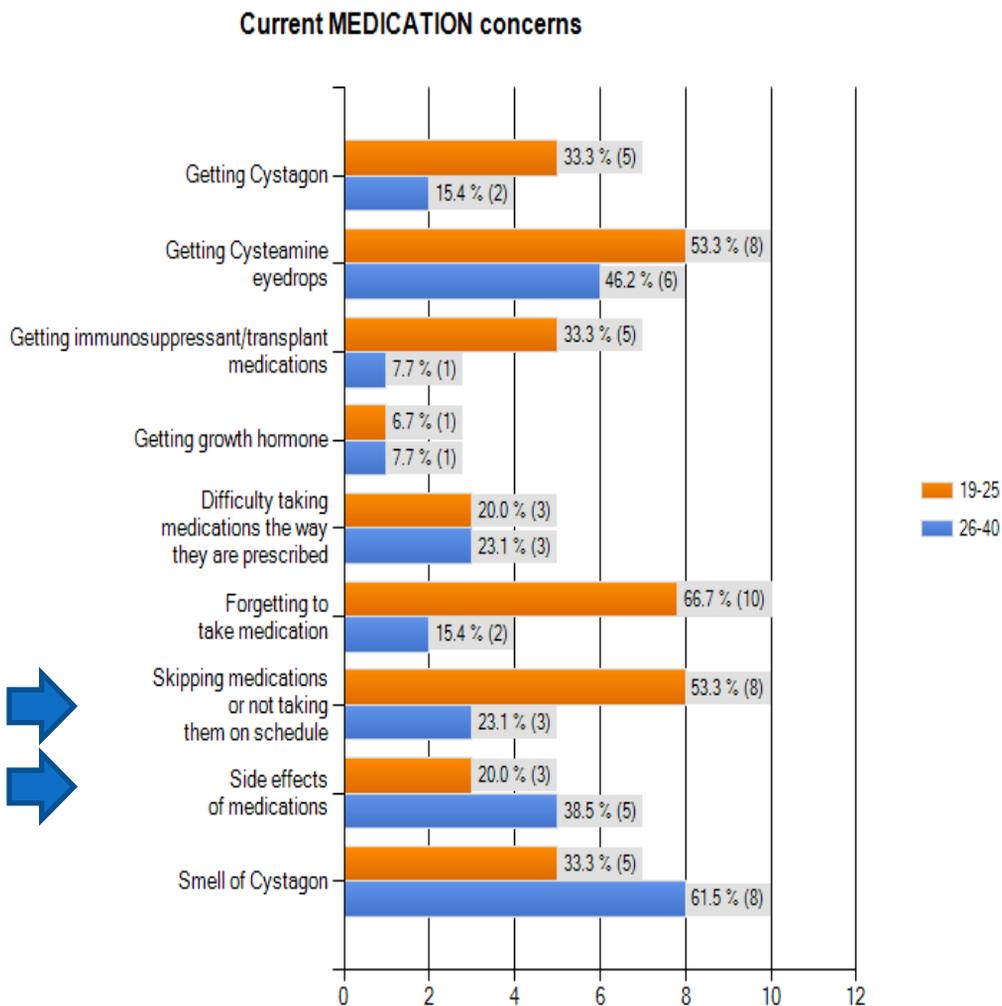


“Muscle wasting is a fairly major issue. There is so much focus on the kidneys, but in reality, once you get a good kidney transplant, this becomes practically a non-issue.”

“My biggest challenge is muscle wasting in the esophagus, diaphragm, hands, and vocal folds. I feel like it is slowly getting worse. I choke on almost all food I eat. It is nearly impossible for me to talk and eat, and I can't talk for about an hour after eating”

While kidney issues are also a primary concern for many adults with cystinosis, respondents wrote eloquently about muscle-wasting and difficulties eating and speaking.

# Medication concerns (adults/emerging adults)



Though the availability and convenience of treatments for cystinosis have improved, adults with cystinosis take on more of the task burden of adhering to their own treatment regimens. Awareness and acknowledgment of these practical and personal challenges, and support to improve and maintain adherence, is crucial. The desire for longer-acting medications was unanimous!

## Lifesaving but intrusive meds: In their own words

"My child's life revolves around medicine, doctors, blood work, and shots.... at this point in his life he gets medicine every 2 to 4 hours."

"We spend so much of our time preparing meds, giving meds, traveling... for appointments that it leaves very little free time to do the things we enjoy."

"...to not have to wake every night to give meds would be wonderful."

"I try not to let it get to me, but it always does every six hours when it's time to take my medication again."

"Maintaining medical stability is very difficult for 2 adults to manage, let alone a child who has cystinosis. Quality of life is the biggest struggle every day because of the rigid, inflexible medication regime and side effects of the medications. It interferes in all aspects of life. Living by the clock is exhausting to manage."

"I would love to actually sleep through the night, considering I've never done that before in my life."

"The only real problem with the cysteamine is getting him up at midnight and at 6 A.M."

"I feel I get left out of a lot of things when I have to stop hanging out with my friends to take my meds... I would like it if the meds would not have to be taken at a rigid time schedule."

## The smell of Cystagon: In their own words

“He smells most of the time like an odor I cannot explain, and school age kids are mean sometimes.”

“[I would like] less smell from Cystagon, [but I am] not complaining. I am happy we have a drug that reduces our complications and allows me to be this healthy.”

“[The smell] makes him feel super self conscious about himself and not very outgoing with others.”

“She's very compliant in taking her meds, although she knows they make her throw up occasionally and that the Cystagon 'stinks' (her word).”

“[I'm] trying different things to control body odor at the moment, such as B2 and chlorophyll gel capsules. I'm convinced the teasing got so bad at school that it was affecting his grades.”

## Hope for longer-acting meds: In their own words

"If the new Cystagon pill, the one where you only have to take it three or two times a day, gets approved then that would be great."

"We are really looking forward to the 12-hour Cystagon."

"[We should be] pressuring [drug companies] to get the 12-hour cysteamine drug to the community as soon as possible. "

"[We need] eye drops [that only] have to be taken one time daily."

"[I would like] to get the 12-hour Cystagon. That's really the only difficult challenge I face right now."

"The main quality-of-life issue is the Cystagon pill. If a more long-term medication could be researched, it would increase my quality of life."

"The development and approval of the RP-103 [12-hour Cystagon] holds great promise in making life easier. Gel eye drop which will last longer in the eyes would also be helpful."

"Eye drops shouldn't take 30 years to approve."

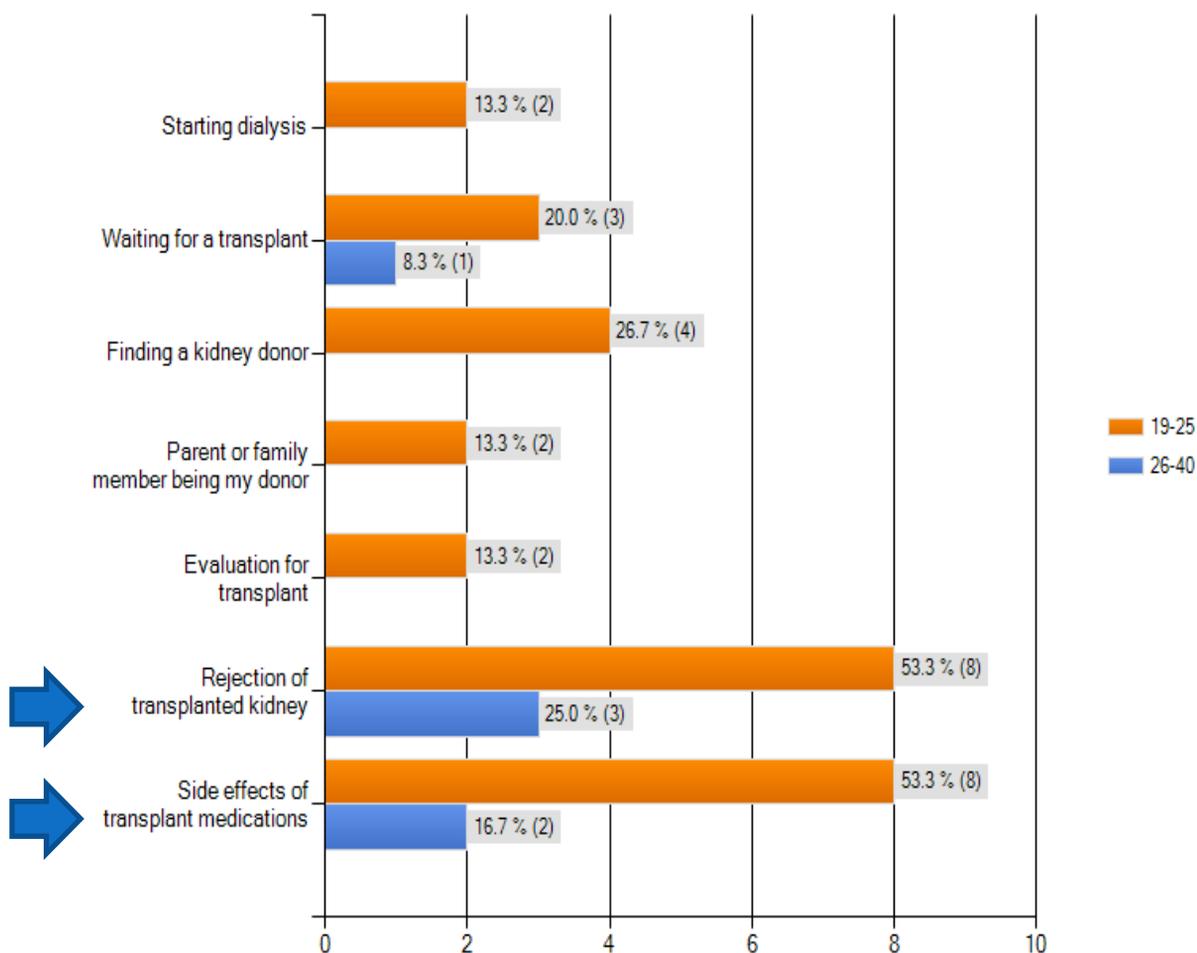
"Once-a-day meds would have a big, positive impact."

"[I would like to see] less medications less often, eye-drop [dosage] reduction, and FDA approval of the eye drops."

"[We need to] improve the quality of medication, a time-release capsule for Cystagon, and/or other medication."

# Dialysis and transplant (adults/emerging adults)

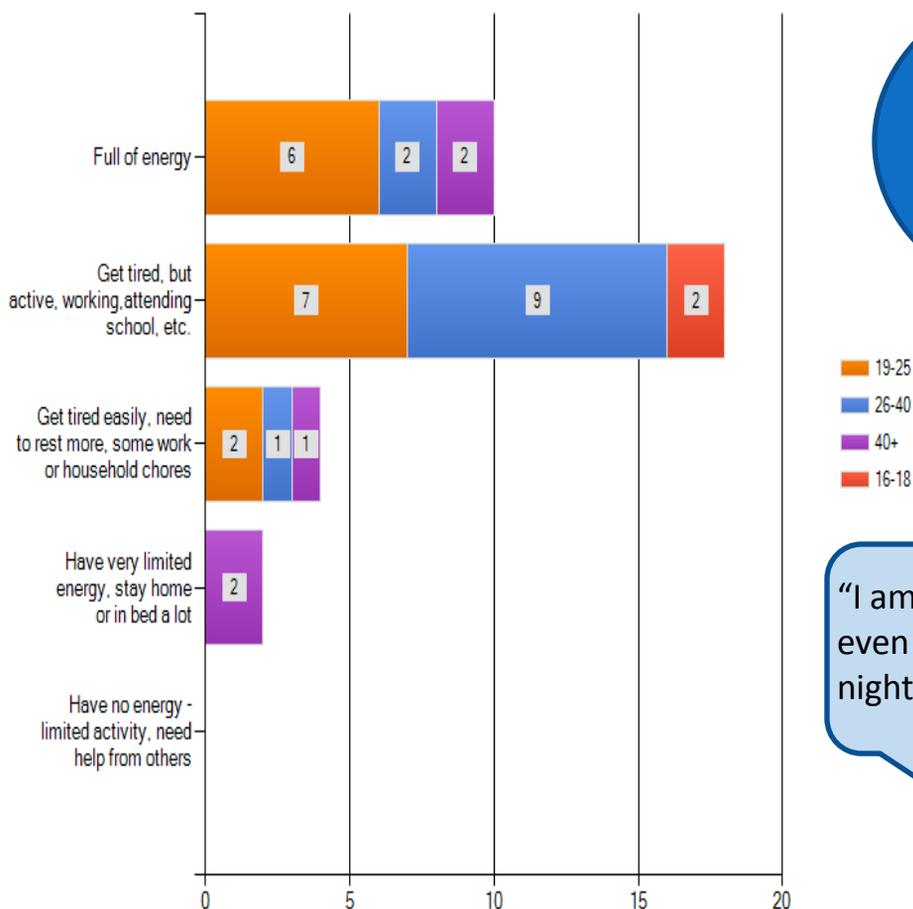
Current concerns about DIALYSIS or TRANSPLANT



Given the nephropathic sequelae of cystinosis and the likelihood of having had a first transplant in early adolescence, adults with cystinosis have ongoing concerns about managing kidney transplants successfully. Ongoing support and education from transplant and nephrology teams may help support adherence, allay fears, and enhance graft survival.

# Energy and fatigue (adults/emerging adults)

How would you describe your health and energy level today



“I am less strong and energetic than most people around me.”

“I am tired all day even with a good night of rest.”

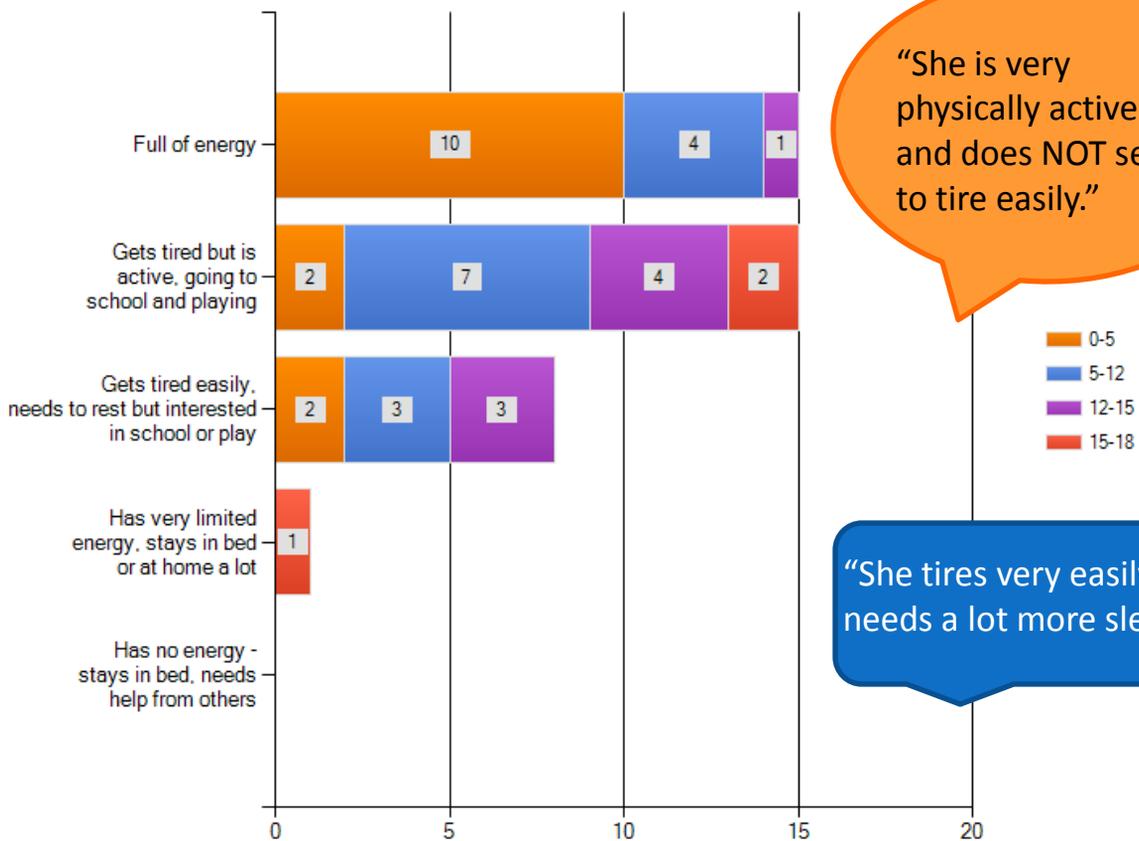
While most adult respondents were “tired, but active,” many wrote about the challenges of staying active while fighting fatigue.

“I feel very tired, but try to push myself.”

“I’m a full-time college student... It’s hard enough doing that without the constant fatigue. I’m tired all the time... it never stops.”

# Energy and fatigue (children)

How would you describe your child's health and energy level today



“She is very physically active and does NOT seem to tire easily.”

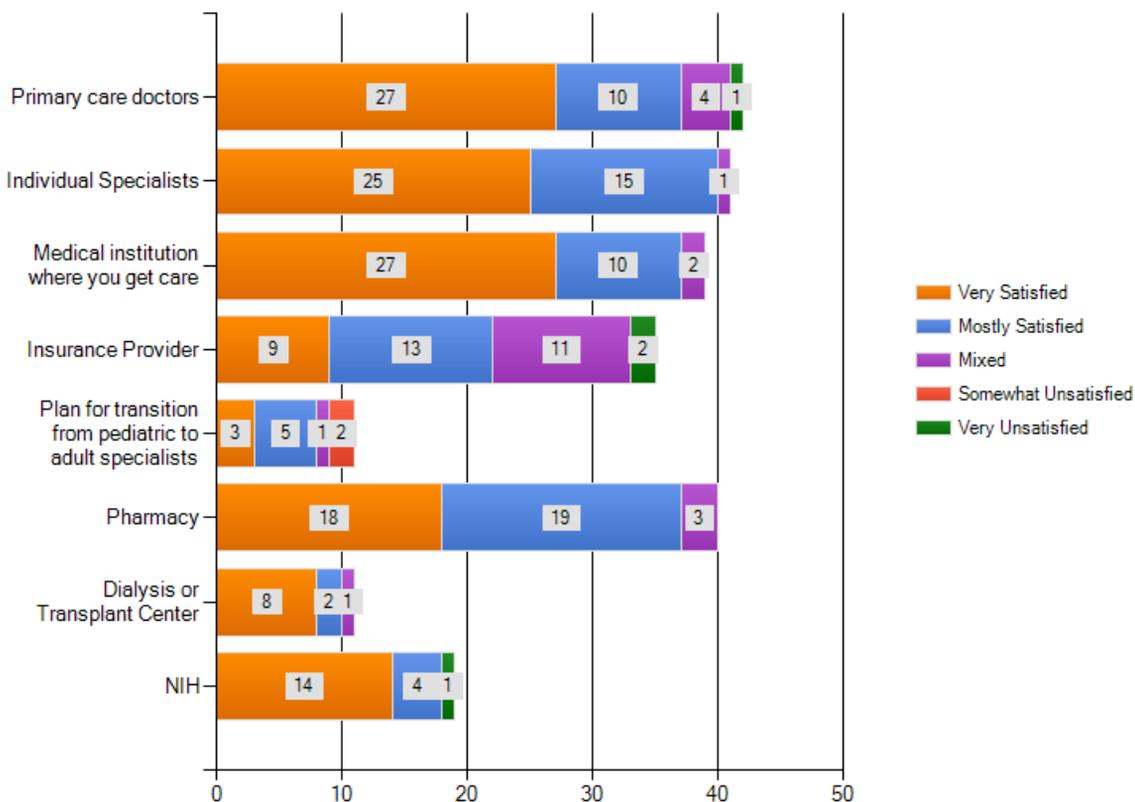
“She tires very easily and needs a lot more sleep.”

“Her energy is not what the average child has.”

“He is a wonderful, normal child who gets in trouble and runs around like a mad man.”

# Satisfaction with health care (families)

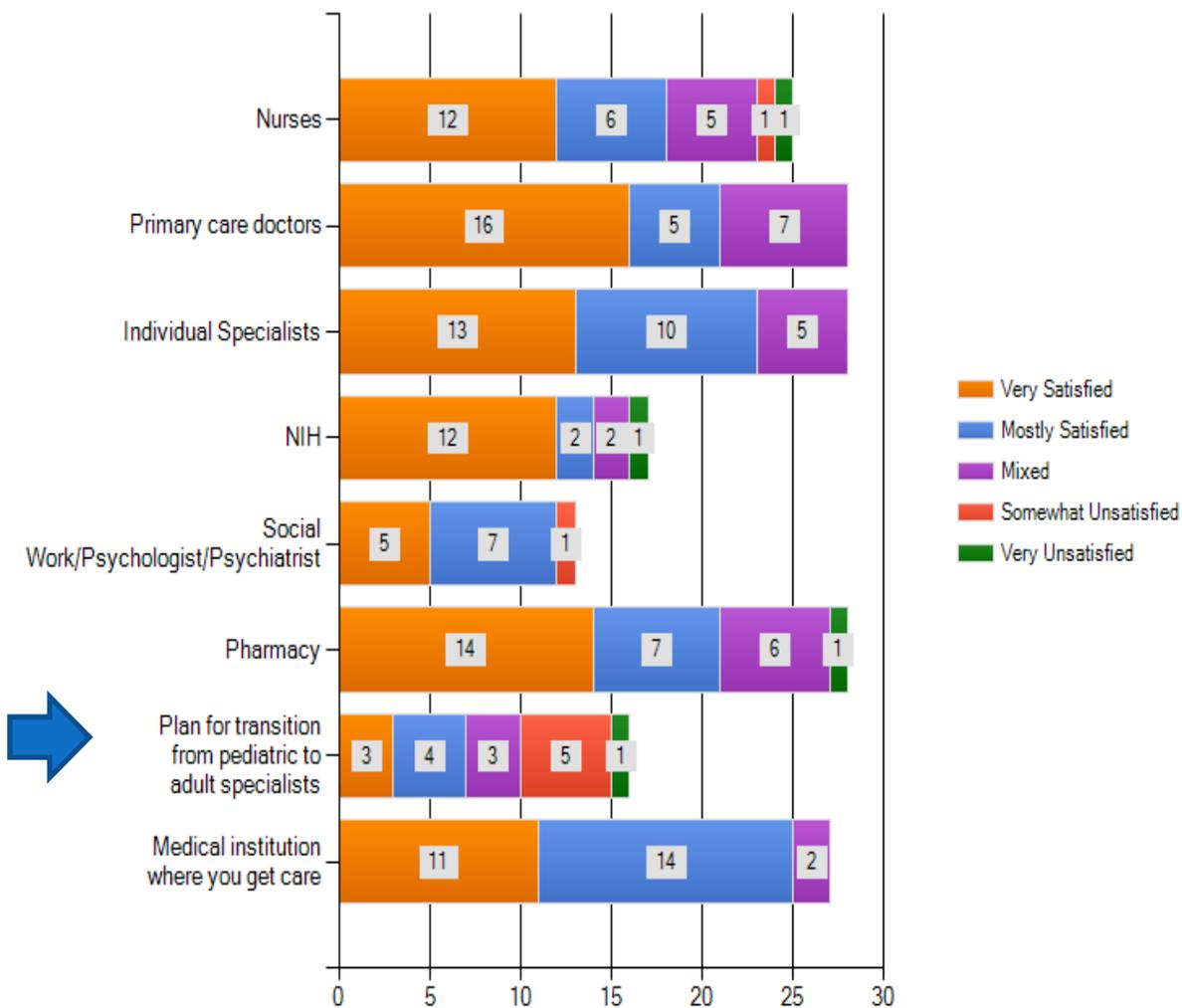
Tell us how you feel about your child's current medical care and services



Although most families expressed satisfaction with their child's health care, concerns about primary care doctors, pharmacies, and insurance plans may be a reflection that these systems lack knowledge about cystinosis, and the hurdles families face in educating those who provided vital services to their children.

# Satisfaction with health care (adults)

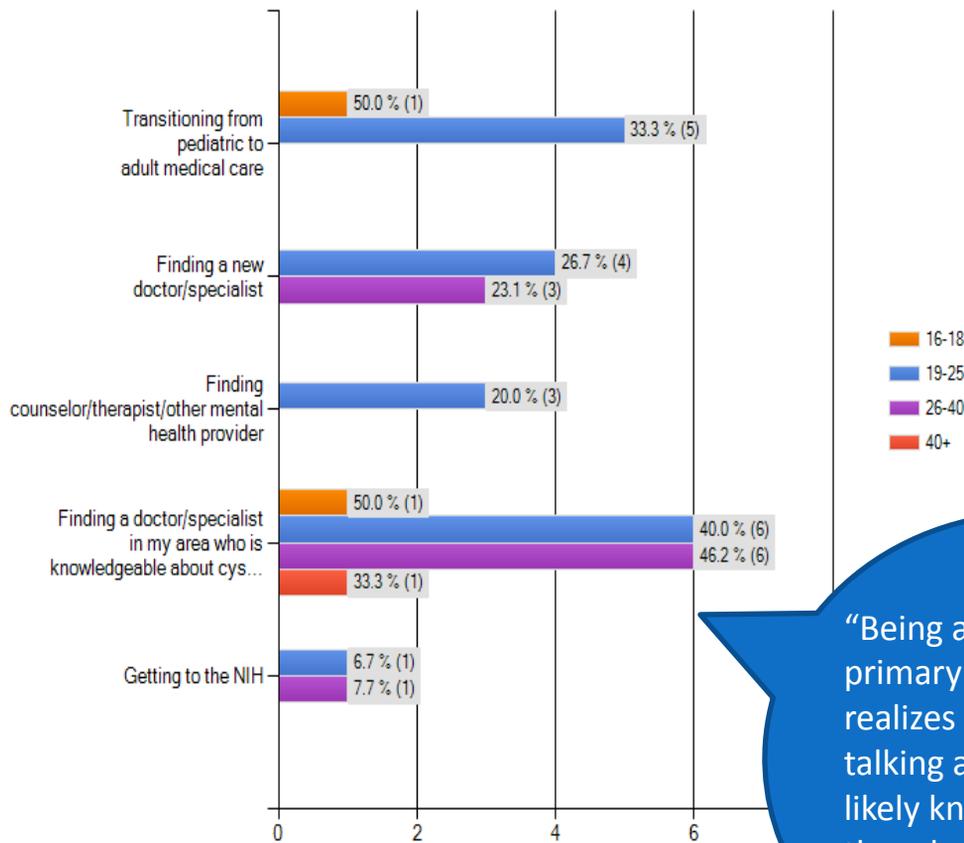
Tell us how you feel about your current medical care and services



While most emerging adults and adults with cystinosis are very satisfied or mostly satisfied with their health care, several respondents expressed dissatisfaction with their provider's plan for transition from pediatric to adult care.

# Health care concerns (adults/emerging adults)

Current concerns about MEDICAL CARE



“Being able to find a primary care person who realizes I know what I'm talking about and most likely know more than they do about how I need to be cared for.”

While “transitioning” typically refers to the move from pediatric to adult care settings, adults with cystinosis will likely change health care providers more than once as jobs and insurance plans change, and may need to provide information to those providers. We are building a library of articles, a transitioning toolkit, and a list of recommended adult specialists on the CRN website.

# Transitioning (adolescents)



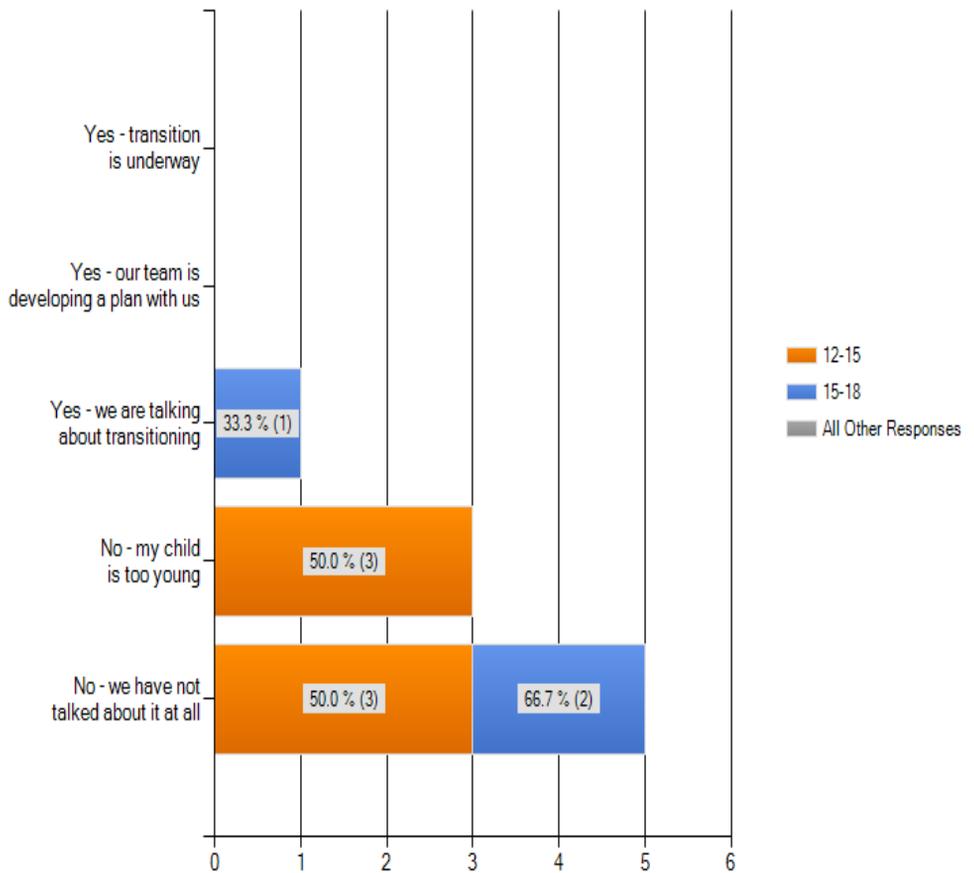
Of the **9** survey respondents with children ages 12-18 with cystinosis:

**1** reported that they were “talking about transitioning”;

**3** families thought their child was too young;

and **5** reported not talking about transitioning at all.

If your child is currently receiving care by one or more pediatric specialists, have you spoken with them about transition to adult care?



Adolescence is the perfect time to develop self-care and independent-living skills. Help your child practice self-care skills early, while you can provide a “safety net.” Encourage your team to start talking directly to your teen, for at least a few minutes every visit. If health care providers aren’t raising these issues, families must!

## Transitioning: In their own words

“The hardest thing I have ever done was to trust my son could manage his own health care needs and step back to let him do it. Sink or swim, his choices, his consequences... we fight all their lives to maximize the quality of their life. 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.*”

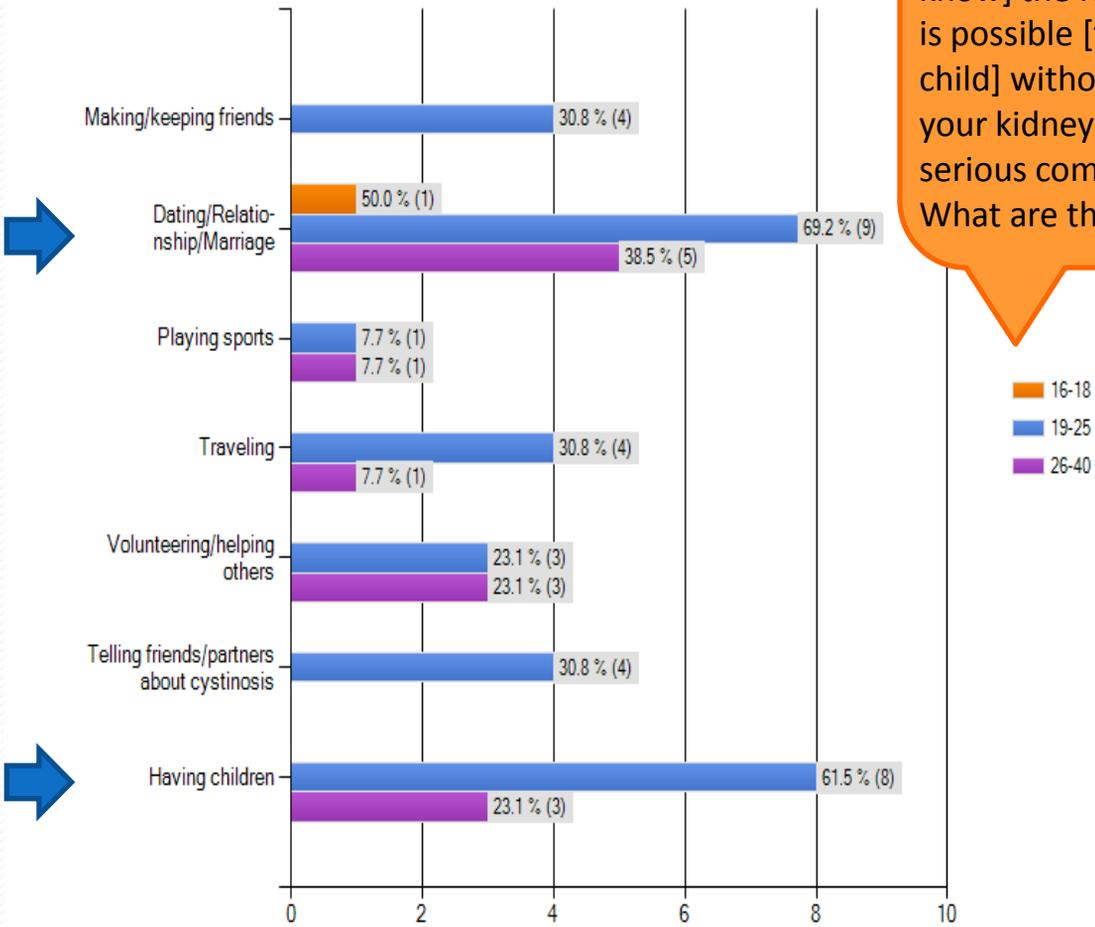
“[It’s important to know] where to go and what to do when you are grown and have to take care of yourself”

“It [transitioning] has been mentioned at annual ‘well-checks,’ but nothing [is] formally being done. At 17, we still have a year and a half of high school. We are beginning to give her more responsibility as far as her care, being more in charge at doctor visits, etc.”

# Social concerns (adults)

“More research and information needs to be out there on childbearing for post-transplant cystinosis patients. [We need to know] the risks and if it is possible [to have a child] without rejecting your kidney and having serious complications. What are the odds?”

Current SOCIAL concerns

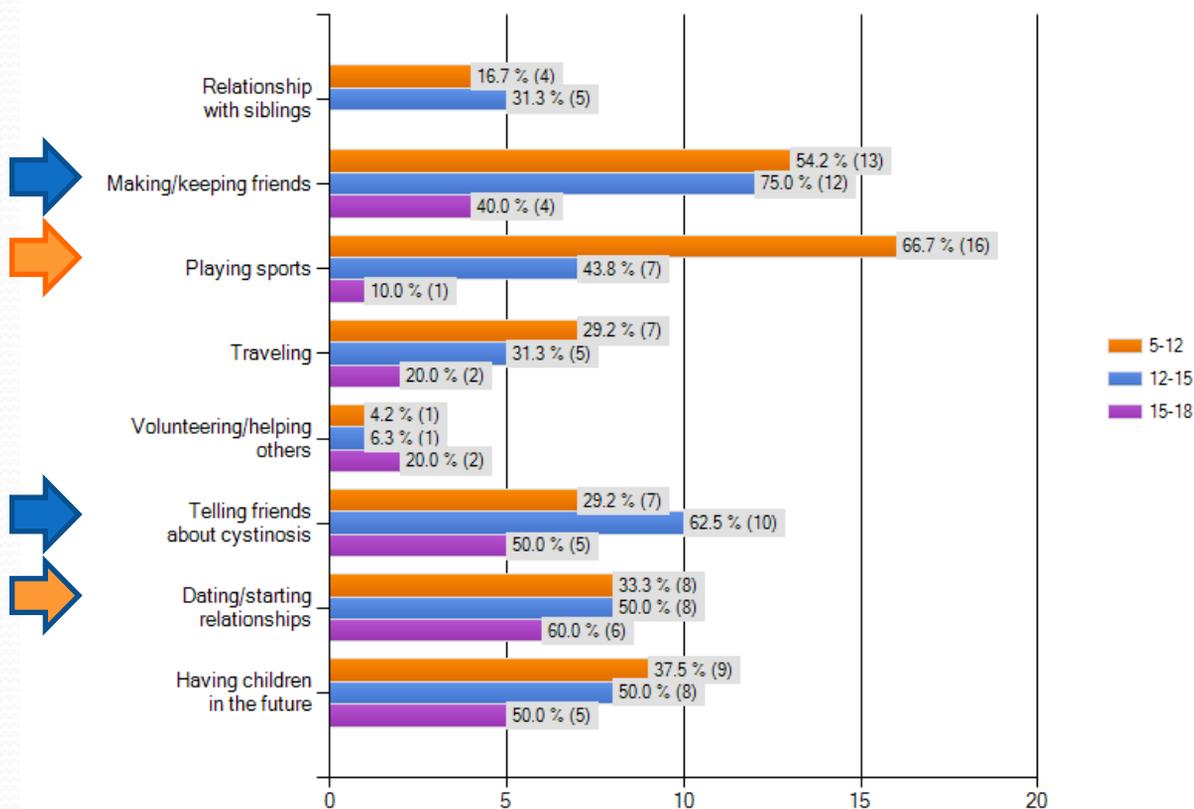


Building relationships and starting families are high priorities for some emerging adults with cystinosis, as they continue to face the stigma of illness, reproductive health challenges, and the potential for early mortality. 73% of men and 29% of women expressed concern about dating, relationships, and marriage, while 41% of men and 29% of women expressed concern about childbearing.

## Social concerns (families)

Families expressed the greatest concern about their child's ability to *make, keep, and manage friendships*. Starting *relationships and families* were of more concern to parents of older children and younger teens, as their child's future may still feel uncertain. The ability to *participate in physical activities* was a major concern for parents of younger children.

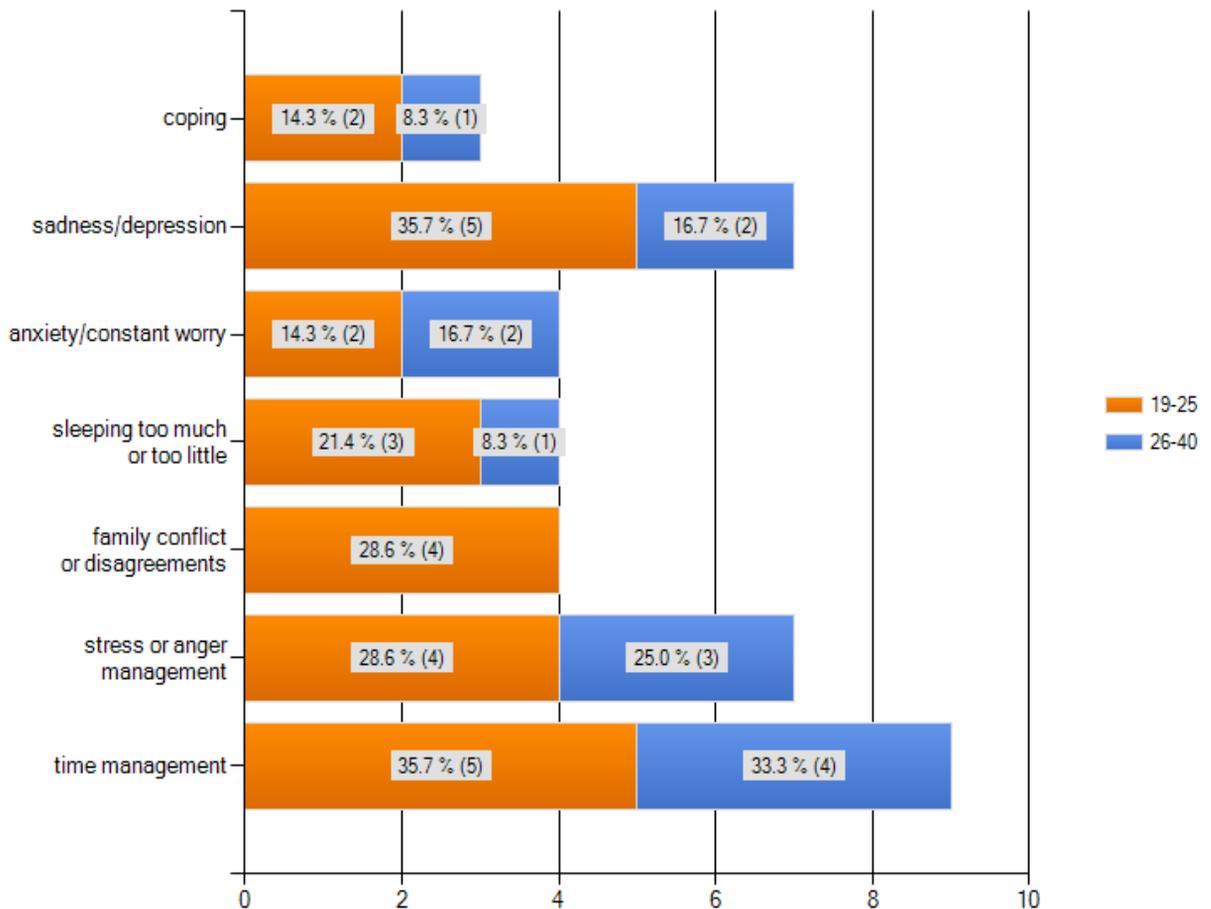
Your Child's Current SOCIAL concerns



“Cystinosis interferes with her ability to do things kids do – ride [a] bike on two wheels, have sleepovers at friends’ houses... Are there activities that benefit children with cystinosis outside of competitive sports? Many cannot keep up with healthier peers after a certain age. Would love more suggestions about art, music, yoga, etc.”

# Emotional/mental health (adults/emerging adults)

Current EMOTIONAL or MENTAL HEALTH concerns:



Sadness and depression, management of anger and stress, and management of time are the most powerful concerns for adults with cystinosis. Concerns with time management may reflect the juggle between day-to-day realities and adherence to one's treatment regimen.

## Ups and downs: In their own words

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 for sure been my amazingly strong support network... Overall I'd say it's been hard, but without all my experiences I wouldn't be me.

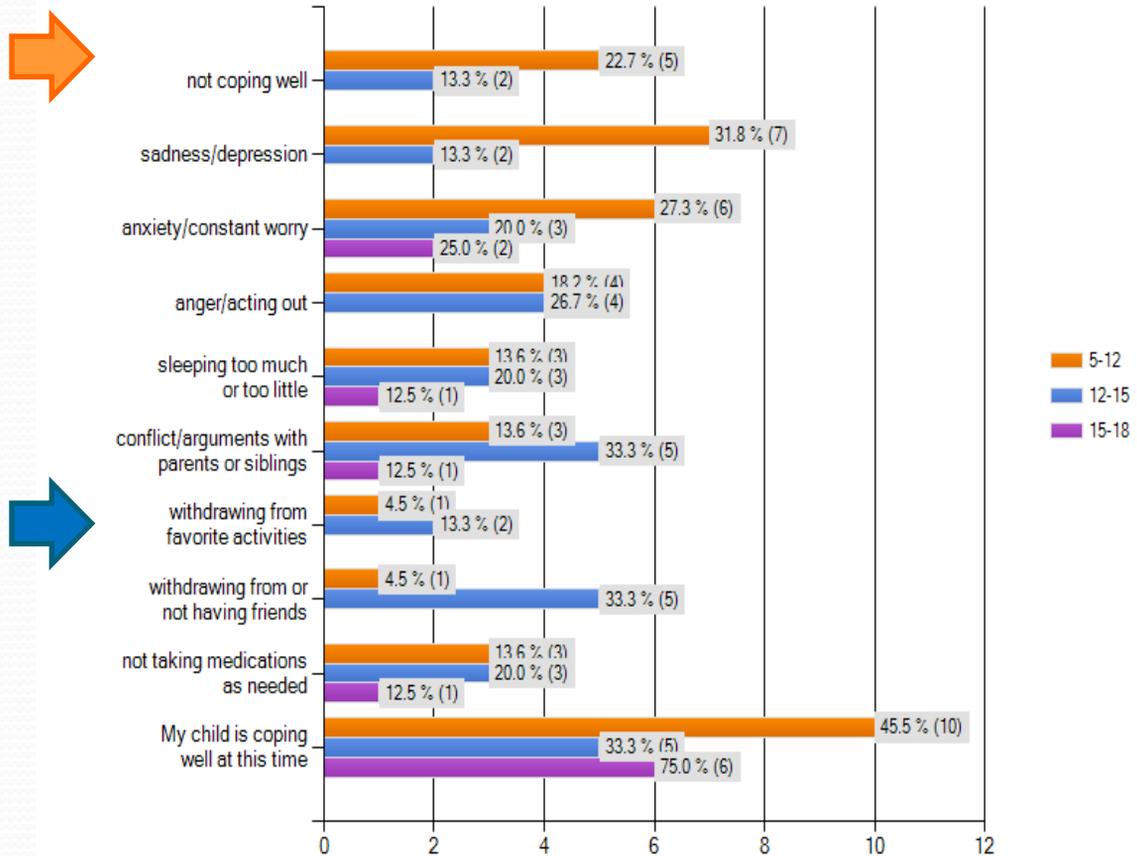
“Generally I'm pretty happy with my life... but it's so much harder than anyone realizes”

It was very hard for me. I was asking why it happened to me, my whole life was falling apart from the day I heard it. I don't know what to say or what to do, all I have to do is to live with cystinosis.

“It 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”

# Emotional/mental health (children)

Your Child's Current EMOTIONAL or MENTAL HEALTH concerns:



While families of younger children expressed concern about their child's individual coping, anxiety, and depression, the families of young teens begin to worry about social and family interactions. Most families, and particularly those with older teens, feel their child is coping well.

## A burden and a blessing for families

How does cystinosis affect your child?	Your Child's age:			
	0-5	5-12	12-15	15-18
<b>Cystinosis is a burden to my child</b>				
Completely true	6	2	3	1
Sometimes true	4	7	3	0
Rarely true	3	3	0	1
Never true for me	0	0	0	1
<b>Cystinosis is a burden to our family</b>				
Completely true	7	5	1	0
Sometimes true	4	4	3	1
Rarely true	1	4	1	1
Never true for me	2	0	1	1
<b>My child is stronger because of cystinosis</b>				
Completely true	4	6	0	0
Sometimes true	4	7	2	3
Rarely true	1	1	3	0
Never true for me	0	0	1	0
<b>My family is stronger because of cystinosis</b>				
Completely true	6	8	0	1
Sometimes true	4	4	4	2
Rarely true	3	1	2	0
Never true for me	0	1	0	0

While some families, particularly those with young children, feel burdened by the disease, most families also feel that living with cystinosis has made them stronger. This likely reflects growing knowledge of the disease and its treatments, greater sources of support, and a more structured approach to coping and to health crises.

“It has made us stronger as a family. It taught our older child patience, tolerance, and unconditional love... It taught our child with cystinosis she is no different; she can be all that she wants to be. It taught my husband and I balance, forgiveness, and has given us an unconditional bond. We embrace each new day as it comes!

## A burden and a blessing for adults

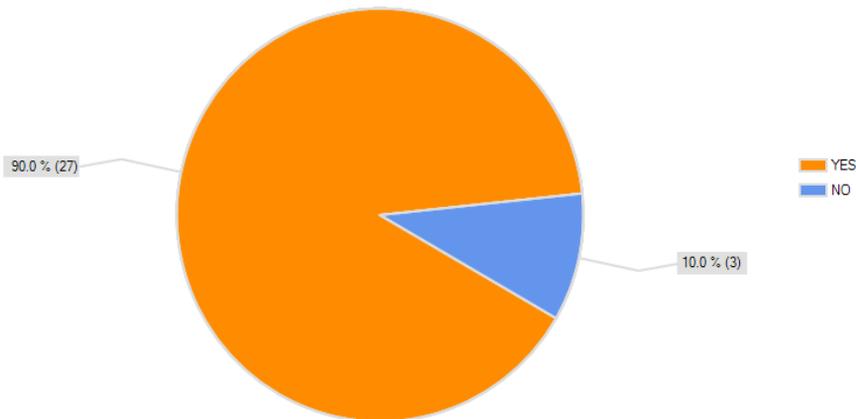
How does cystinosis affect you?	Your age:	
	19-25	26-40
<b>Cystinosis is a burden to me</b>		
Completely true	2	0
Sometimes true	4	7
Rarely true	5	2
Never true for me	2	3
Not Applicable to me	1	1
<b>Cystinosis is a burden to my family</b>		
Completely true	3	1
Sometimes true	8	5
Rarely true	2	2
Never true for me	0	4
Not Applicable to me	1	1
<b>I am stronger because of cystinosis</b>		
Completely true	7	4
Sometimes true	5	5
Rarely true	2	2
Never true for me	1	0
Not Applicable to me	0	1
<b>My family is stronger because of cystinosis</b>		
Completely true	6	5
Sometimes true	4	3
Rarely true	3	3
Never true for me	0	0
Not Applicable to me	1	1

“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.”

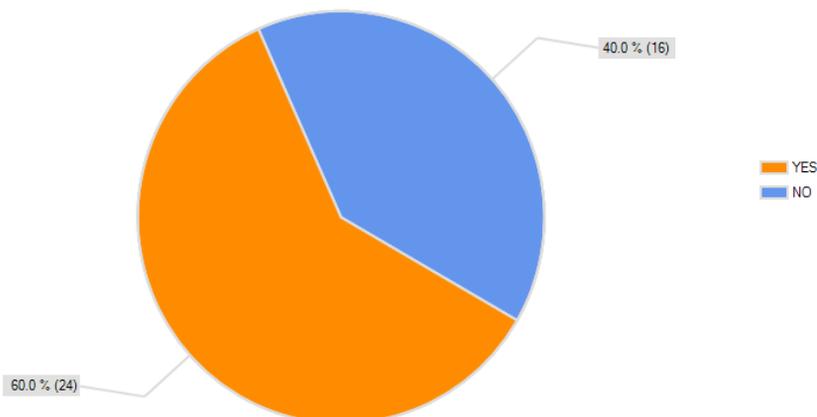
Emerging adults, more than any other group, said that it was *rarely true* that cystinosis was a burden, and most felt they and their families were stronger because of their experience. While this may reflect an optimistic bias, it also suggests a group of individuals who have integrated cystinosis into their everyday lives in a successful way.

# Research participation

Have you ever been a participant in a research study about cystinosis or treatments for cystinosis?



Have your child ever been a participant in a research study about cystinosis or treatments for cystinosis?



This is a truly pro-active community. 90% of adult respondents and 60% of families report currently or previously participating in research studies about cystinosis or treatments for cystinosis.

# Research priorities for families and adults

