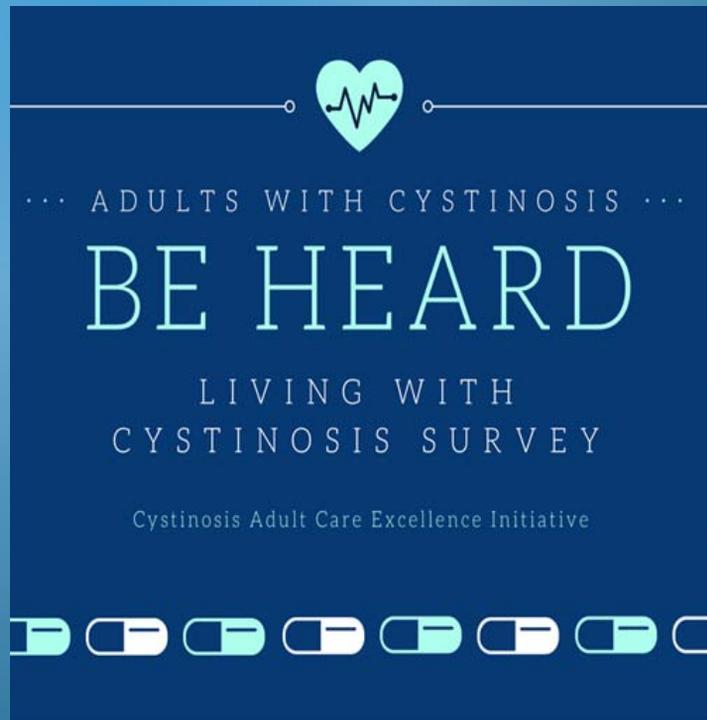


Cystinosis Adult Care Excellence (ACE) Initiative

Initial Survey Results and
Recommendations
February 2016



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A Moment For Action

- We are extremely grateful for the incredible progress made in understanding and treating cystinosis, and the many individuals with cystinosis who selflessly participated in research studies and experimental treatments in support of this work. These efforts have both extended and improved the lives of children and adults living with cystinosis.
- However, adults now living with cystinosis are facing multiple medical and psychosocial challenges. Some adults have had the benefit of early diagnosis and treatment, while others have not. Many adult patients are dying prematurely, due to long-term complications of cystinosis.
- The cystinosis community must make key investments of resources and time to identify the shifting needs of the adult population, to empower these individuals, and to ensure treatment is available to meet their needs. Action taken now can make a difference in their ability to live longer and healthier lives, and improve the future for younger patients.

“Many of the adults are experiencing huge health issues, and I believe more research is needed to improve the lives of these adults. I hope from small beginnings this group will expand to include adults from all over the world working together for better treatments.” – Sue Scott, ACE workgroup member

A Moment For Action

- The goal of the Adult Care Excellence (ACE) Initiative is not to dwell on the negative, but to highlight the work that still needs to be done. We are incredibly grateful to the organizations and researchers who have worked so diligently on behalf of our cystinosis community.
- Cystinosis continues to present immense challenges which can foreshorten lives, and which may result in discomfort, disability, and distress.
- The ACE Initiative Working Group was formed in September 2015 with the goal of empowering adults with cystinosis, and to give voice to their specific needs and concerns. Our aim is to develop priorities for improved treatments, research, education, support, and advocacy for adults living with cystinosis.
- The working group consists of adults with cystinosis, family members, researchers, and clinicians dedicated to strengthening our understanding of the challenges that exist and the resources needed for living with cystinosis in adulthood. The Working Group is not directly linked to any advocacy organization, research institution, or corporate entity.
- We are pleased to present to you the preliminary results of the Living with Cystinosis Surveys and the recommendations of the Cystinosis ACE Initiative on Rare Disease Day 2016. Thank you to everyone who participated in and assisted in designing and distributing the surveys and organizing this report. We hope the information collected here will move us closer to our goal of empowering and improving quality of life for adults with cystinosis.

Please contact us at CystinosisACE@gmail.com with any questions or concerns. We welcome collaboration with all interested individuals. We continue to accept completed surveys in an effort to include as many voices as possible.

The Living with Cystinosis Surveys

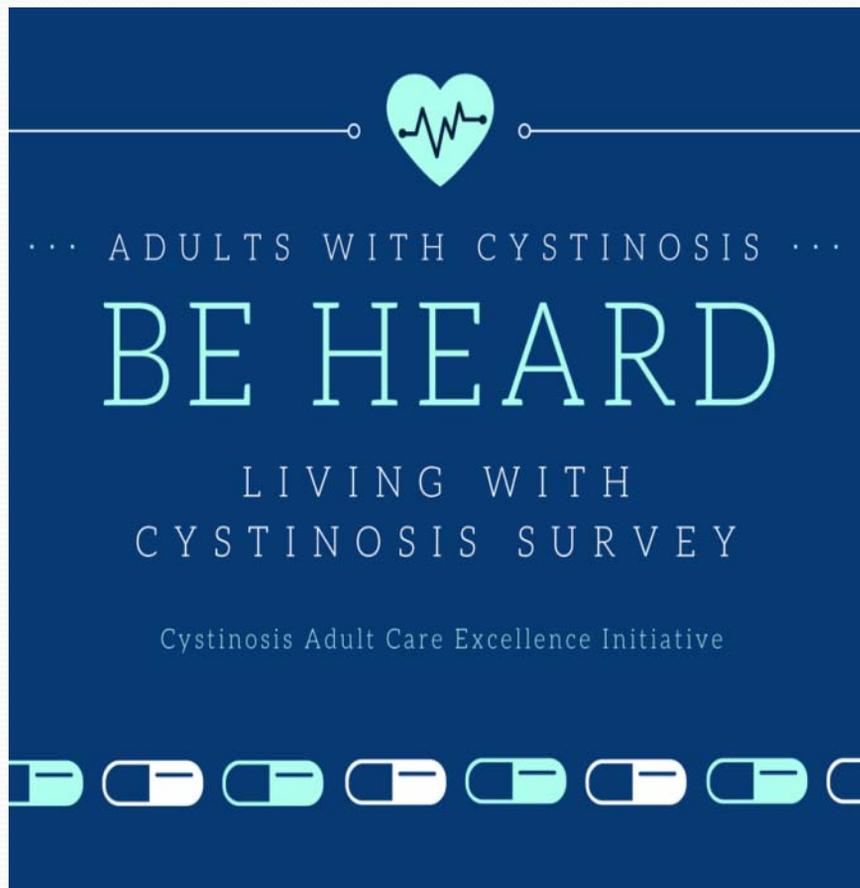
- In December 2015, the ACE Initiative Working Group developed and launched an online survey to inquire about the needs and concerns of adults (age 18+) with cystinosis, titled "Living with Cystinosis 2015 / 2016" (referred to here as Survey 1). Our goal was to give a collective voice to adults with cystinosis. You can view the survey questions here: <http://svy.mk/1lpkJqi>.
- After launching the Living with Cystinosis 2015 / 2016 survey, we noted that participants were adding many open-ended comments regarding health concerns. In an effort to learn more about the specific health concerns of adults living with cystinosis, in January 2016, we launched a second survey, "Living with Cystinosis Survey: Your Health" (referred to here as Survey 2). You can view the survey questions here: <http://svy.mk/1QTuz1v>.
- The Living with Cystinosis 2015 / 2016 survey was adapted from the 2011 Living with Cystinosis survey, which focused on patients of all ages and their parents⁴. The survey includes both multiple choice and open-ended questions. Thus far, 94 responses have been analyzed for Survey 1, and 52 responses have been analyzed for Survey 2. These English-language surveys garnered responses from around the world. Translation of Survey 1 to other languages is under way.

The Living with Cystinosis Surveys

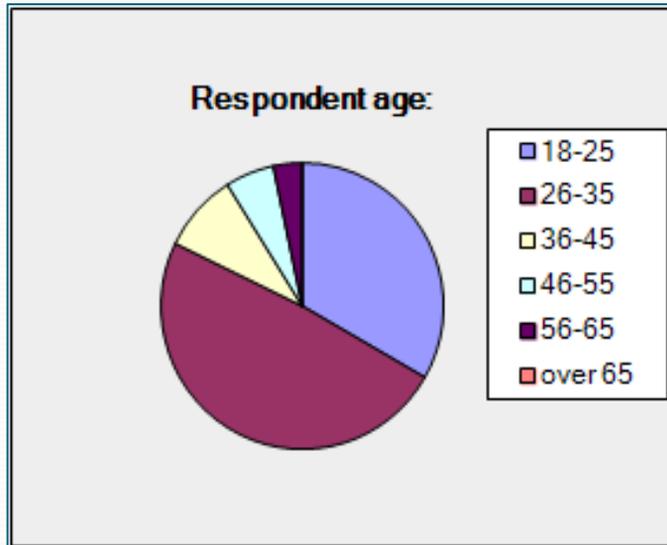
- The survey results help strengthen our understanding of issues of concern to adults living with cystinosis, and provide details of health and medication challenges. In this presentation, the voices of adults living with cystinosis are heard through selected quotes taken from answers to open-ended questions within the surveys.
- The results of both surveys and Working Group recommendations related to the areas of treatment, research, education, support, and advocacy will be shared on the following pages.

Thank you to all of the adults with cystinosis who participated in the surveys. Your efforts will continue to pave the way for improved understanding and treatments.

The Living with Cystinosis Surveys Results – February 2016



Age & Geographic Location – Survey 1 (Q2)



The first Living with Cystinosis Survey received 94 responses. Respondents included adults with cystinosis ranging in age from 18 to greater than 56 years of age. While most responses were from the United States and United Kingdom, we also received responses from Australia, New Zealand, Canada, Ireland, Brazil, Poland, Germany, Iran, and Mexico.

Age & Geographic Location – Survey 1 (Q2)

Current Age		
18-25	33.3%	30
26-35	48.9%	44
36-45	8.9%	8
46-55	5.6%	5
56-65	3.3%	3
over 65	0.0%	0
answered question		90
skipped question		4
Gender		
Female	60.0%	54
Male	40.0%	36
answered question		90
skipped question		4

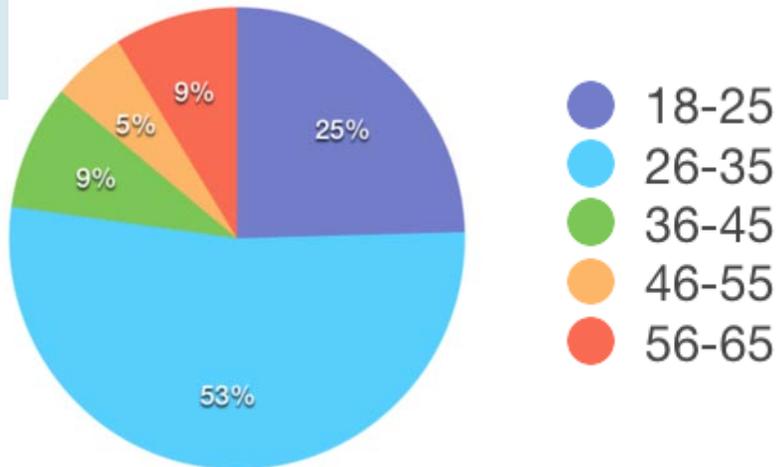
Country		
USA	60.7%	34
UK	14.3%	8
Australia	5.4%	3
Canada	5.4%	3
Ireland	3.6%	2
Brazil	1.8%	1
Poland	1.8%	1
Germany	1.8%	1
Iran	1.8%	1
Mexico	1.8%	1
answered question		55
skipped question		39

Education & Employment – Survey 1 (Q6&8)

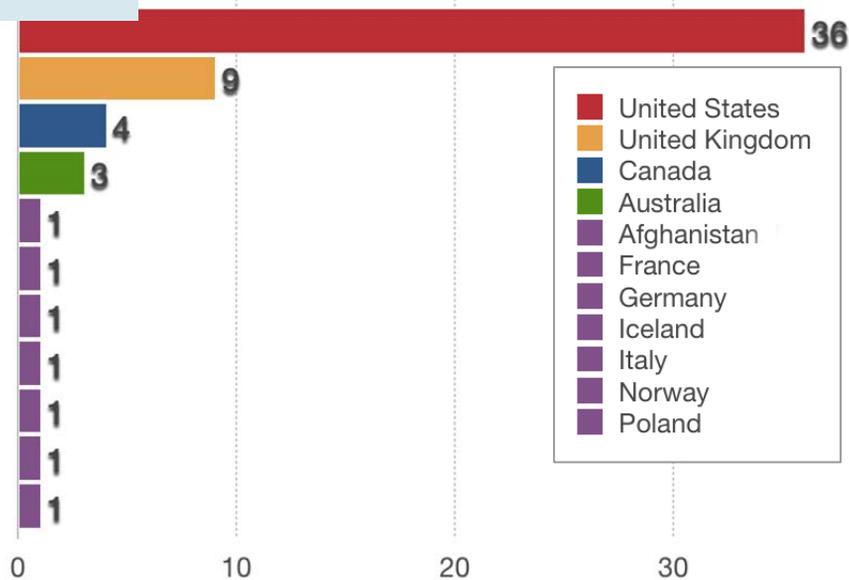
Level of Education completed		
Elementary/Middle School	11.6%	10
High School	41.9%	36
Vocational/Trade School	10.5%	9
College - Associate's Degree	16.3%	14
College - Bachelor's Degree	23.3%	20
Master's Degree or higher	12.8%	11
Current level of employment:		
Student	7.2%	6
College Student	7.2%	6
Working part-time	24.1%	20
Working full-time	37.3%	31
Volunteering	7.2%	6
Unemployed	13.3%	11
Receiving Disability (SSI or SSDI) (USA & Canada))	24.1%	20

Age & Geographic Location – Survey 2 (Q3&5)

AGE



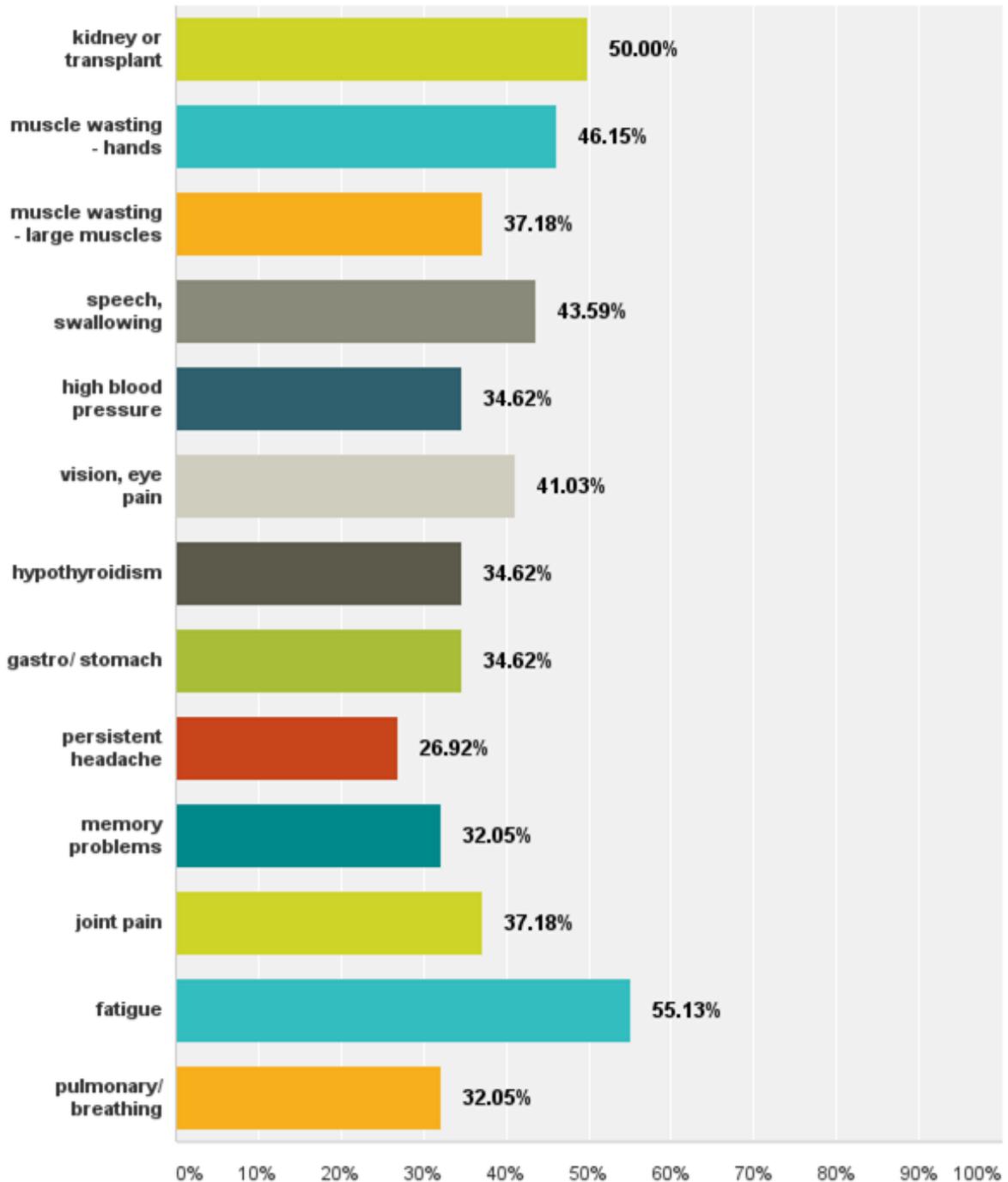
LOCATION



The second survey, Living with Cystinosis - Your Health, received 52 responses. Respondents included adults with cystinosis ranging in age from 18 to greater than 56 years of age. While most responses were from the United States and United Kingdom, we also received responses from Australia, Canada, France, Iceland, Italy, and Poland.

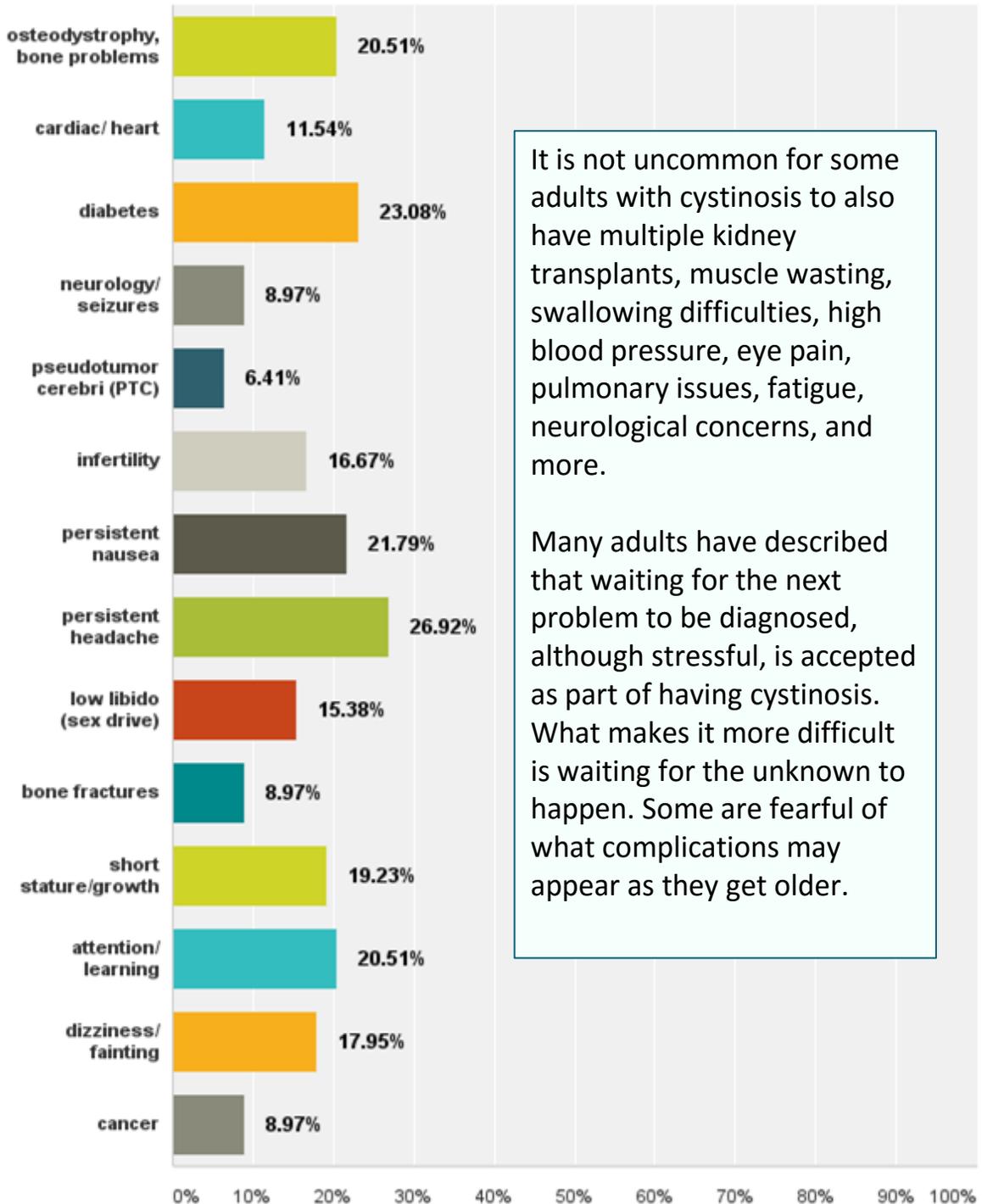
Medical Concerns – Survey 1 (Q14)

Answered: 78 Skipped: 20



Medical Concerns – Survey 1 (Q14, cont.)

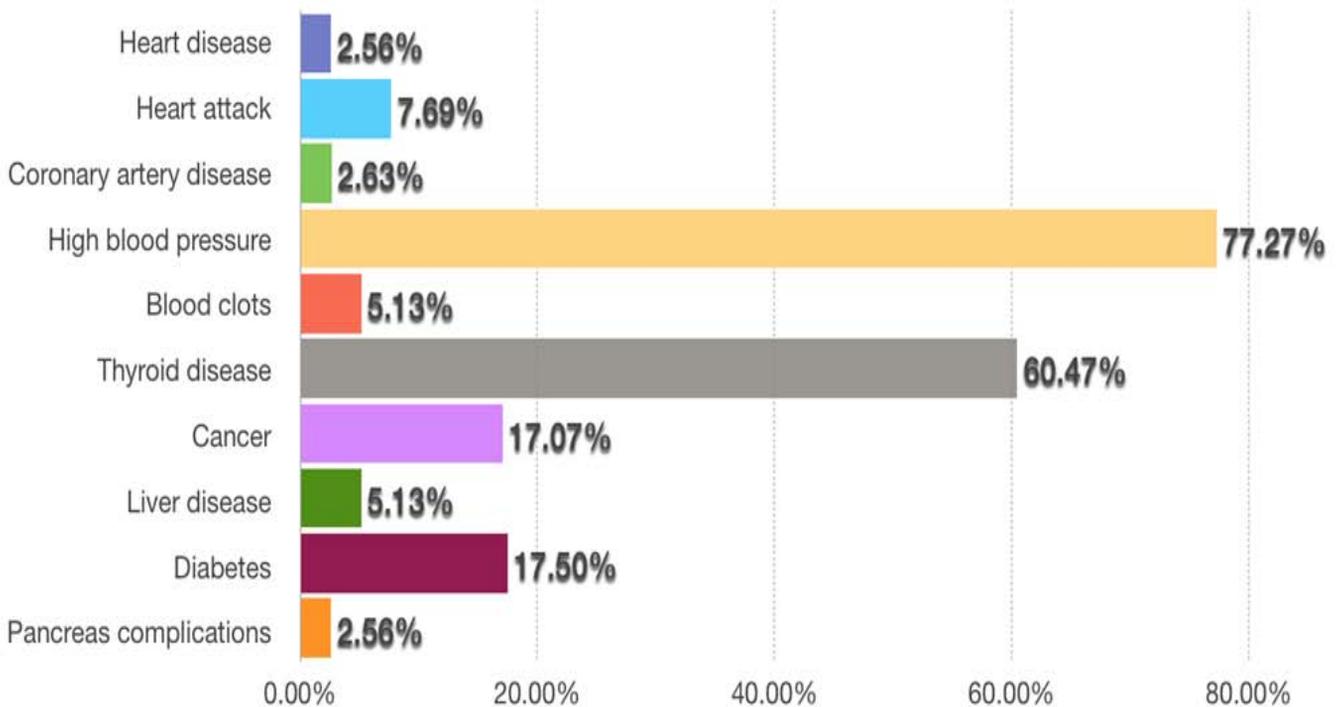
Answered: 78 Skipped: 20



Medical Concerns – Survey 2 (Q18)

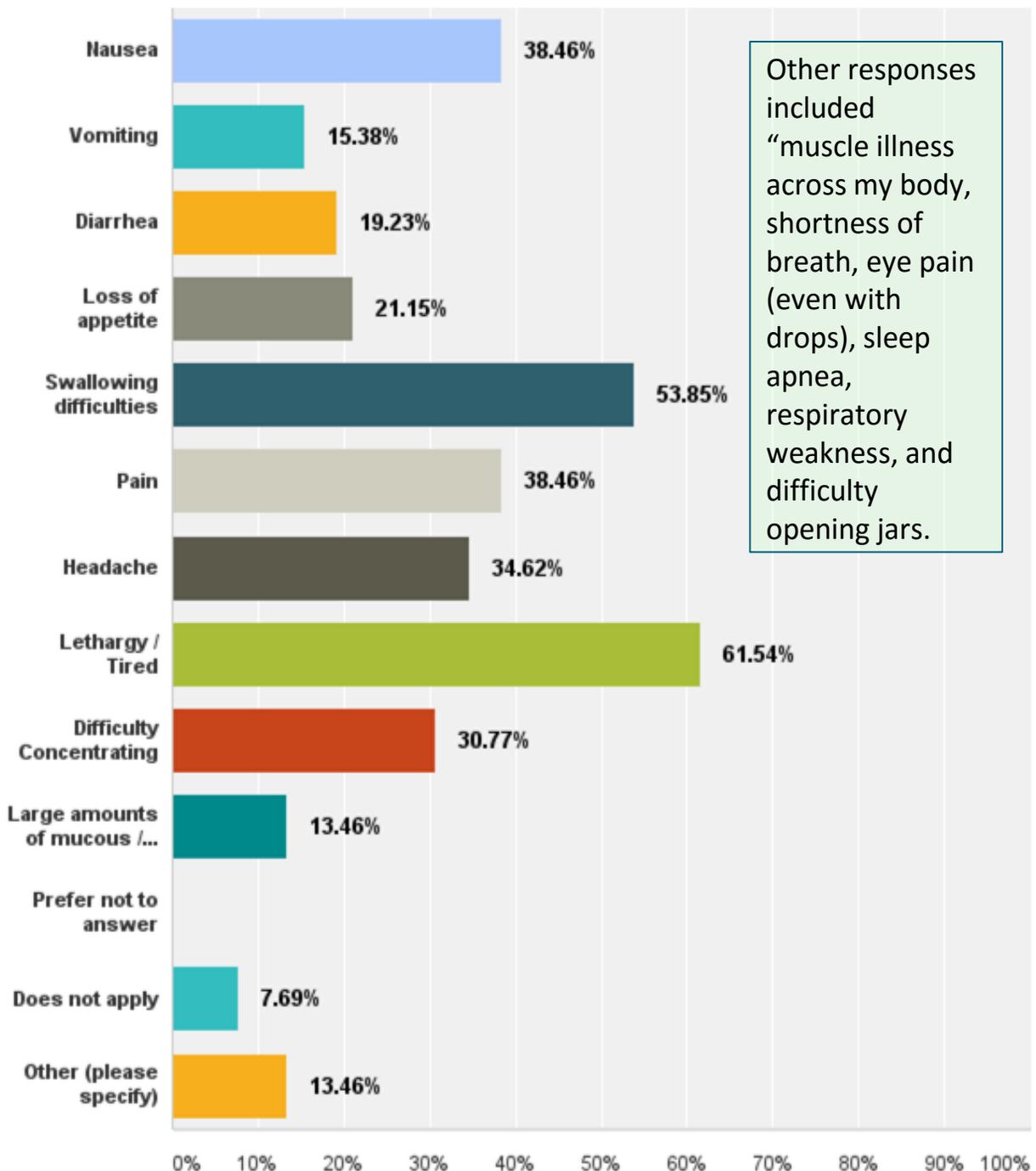
Have you experienced any of the following medical conditions?

Answered: 46 Skipped: 6



Frequency of Cystinosis-Related Symptoms (occurring at least once weekly) – Survey 2 (Q11)

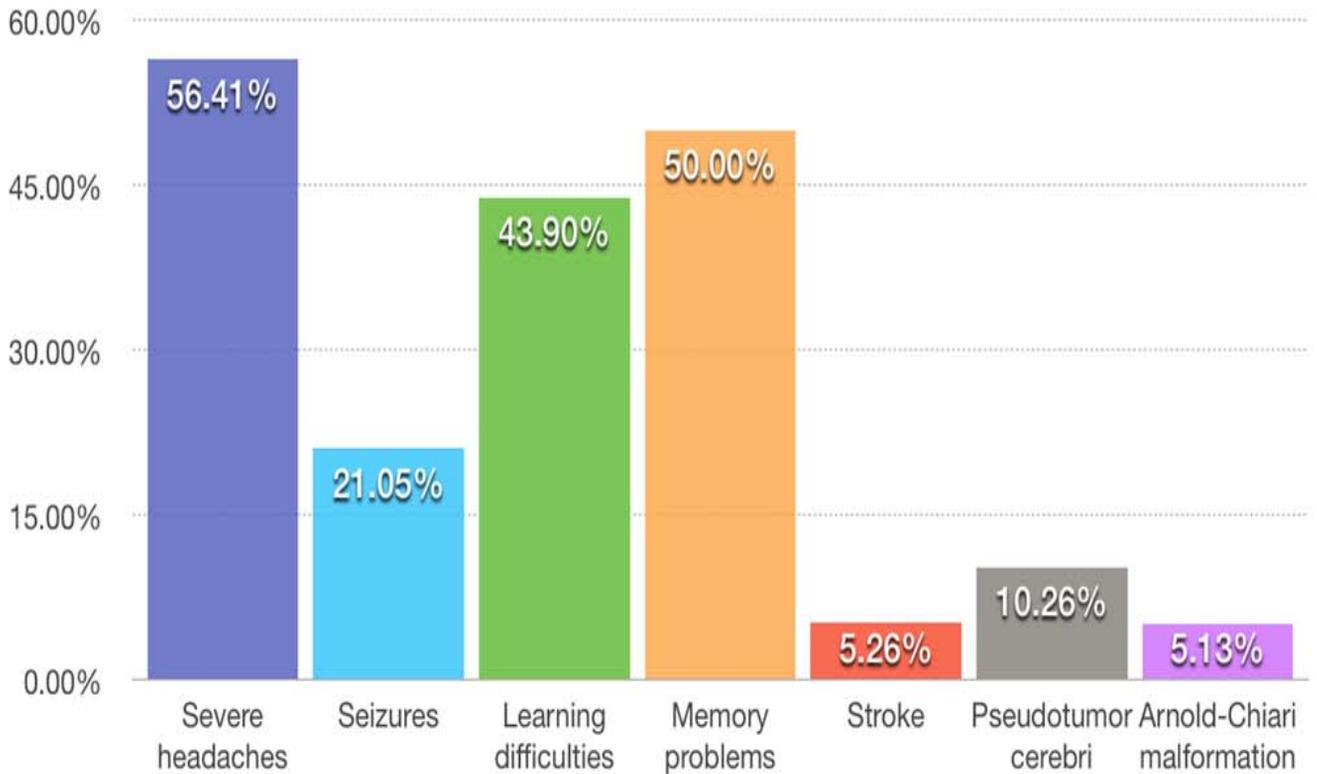
Answered: 52 Skipped: 0



Neurological Health – Survey 2 (Q19)

Have you experienced any of the following neurological conditions?

Answered: 41 Skipped: 11



Respiratory & Pulmonary Health – Survey 2 (Q20)

Have you experienced any of the following respiratory/pulmonary/breathing complications?

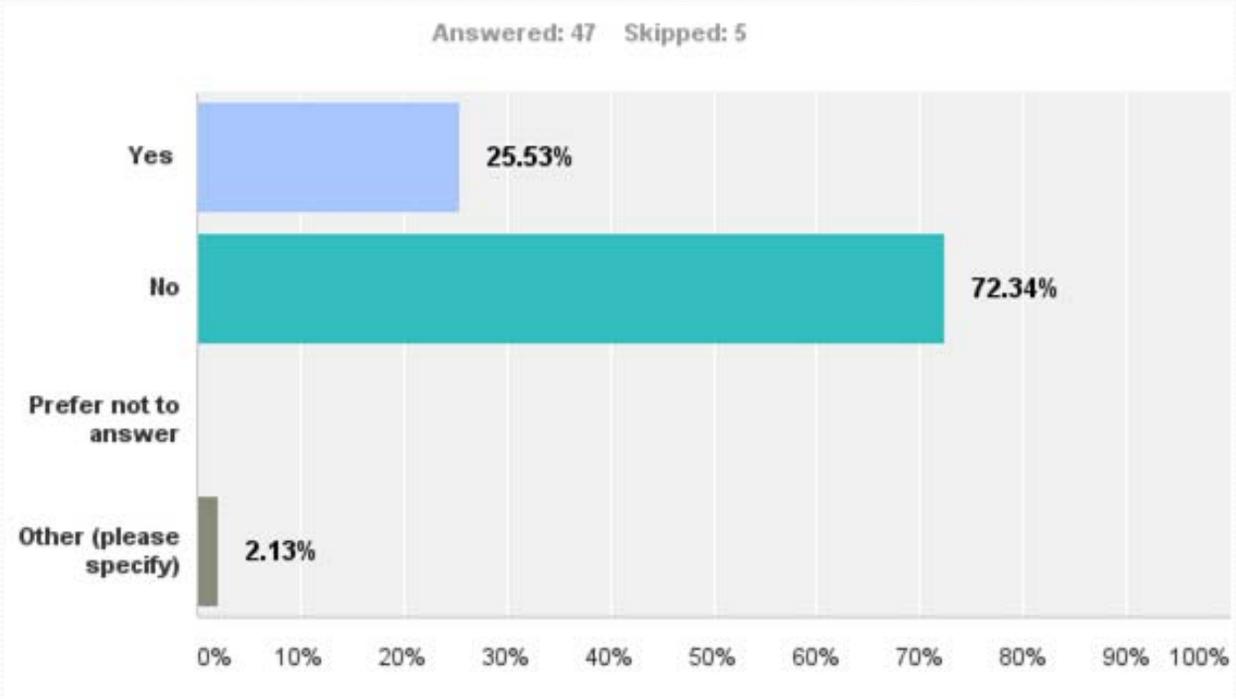
“Occasionally need oxygen support. Occasionally need BIPAP during the day, particularly during upper respiratory infections.”

Answer Choices	Responses
Does not apply	48.94% 23
I had pulmonary function tests	29.79% 14
I experience gastro esophageal reflux	17.02% 8
I use a Bipap or Cpap device to assist with breathing at night?	10.64% 5
I am short of breath at rest or with minimal activity	10.64% 5
I experienced aspiration	8.51% 4
Other (please specify)	8.51% 4
I see a lung disease specialist	6.38% 3
I need the support of CPAP/BIPAP during upper respiratory infections	4.26% 2
I have been treated for aspiration pneumonia	4.26% 2
I am dependent on CPAP/BIPAP during the daytime, if yes how many hours?	2.13% 1
I wake at night coughing from gastro esophageal reflux	2.13% 1
I have been hospitalized for respiratory failure	2.13% 1
I have participated in a Pulmonary Rehab program	2.13% 1
I have been instructed by a pulmonologist to spot-check your oxygen saturation	2.13% 1
I am dependent on others for help due to your respiratory weakness	2.13% 1
I am on oxygen therapy	0.00% 0
Prefer not to answer	0.00% 0
Total Respondents: 47	

“...short of breath climbing stairs, moving up or down quickly.”

Voice Health – Survey 2 (Q21)

Do you have difficulties with voice weakness?



“I think it is mild right now. I manage very well.”

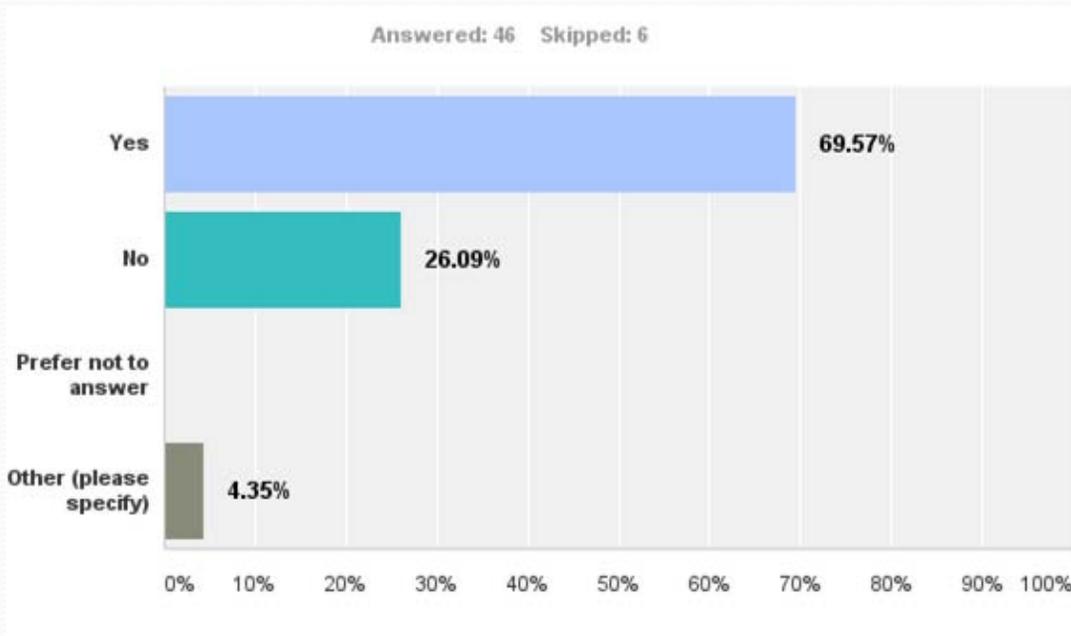
“I run out of breath after very short sentences.”

“I stopped singing in choir because I would lose my voice after practice.”

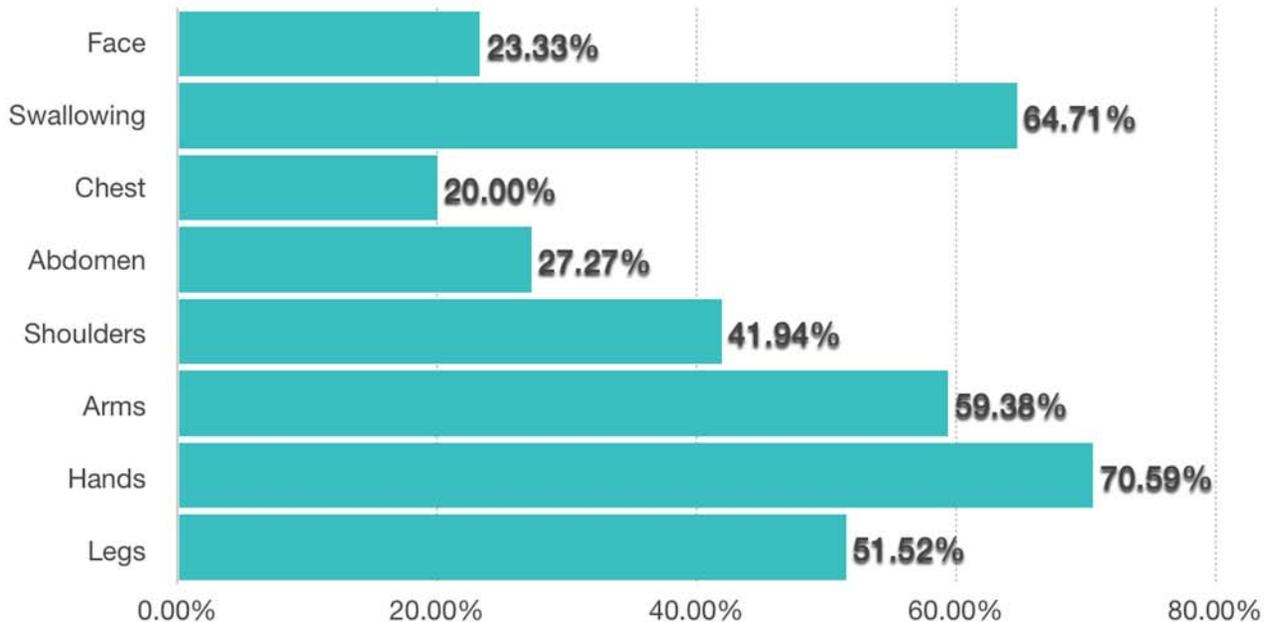
“I have to repeat myself a lot. It gets frustrating.”

Muscle Health – Survey 2 (Q26&27)

Do you experience muscle weakness? Where?



Answered: 37 Skipped: 15



Swallowing Health – Survey 2 (Q30)

Answer Choices	Responses	
Difficulty swallowing pills	32.61%	15
Sensation of food sticking in the throat or chest	32.61%	15
Frequent throat clearing or coughing or both after the swallow	30.43%	14
Coughing or choking with food or liquid or both	26.09%	12
Prefer not to answer	21.74%	10
Difficulty moving food or liquid or both out of the mouth and into the throat	17.39%	8
Difficulty getting the swallow started	17.39%	8
Burping during or after or both meals	15.22%	7
Foreign body sensation in throat	15.22%	7
Coughing or choking on saliva during non-mealtimes	13.04%	6
Needing to avoid certain food or liquid or both	10.87%	5
Waking at night coughing or choking	10.87%	5
Difficulty chewing	8.70%	4
Regurgitation or being unable to keep food or liquid or both down	8.70%	4
Pain during swallow	6.52%	3
Nose running during meals	6.52%	3
Large amounts of saliva / mucous	6.52%	3
Does not apply	6.52%	3
Losing food or liquid or both from your mouth during meals	4.35%	2
Food or liquid or both coming out of the nose	4.35%	2
Sneezing during meals	2.17%	1
Eyes watering during meals	2.17%	1
Sudden coughing after lying down	2.17%	1
Keep food in cheeks until able to swallow	2.17%	1
Total Respondents: 46		

Swallowing Concerns – In Their Own Words

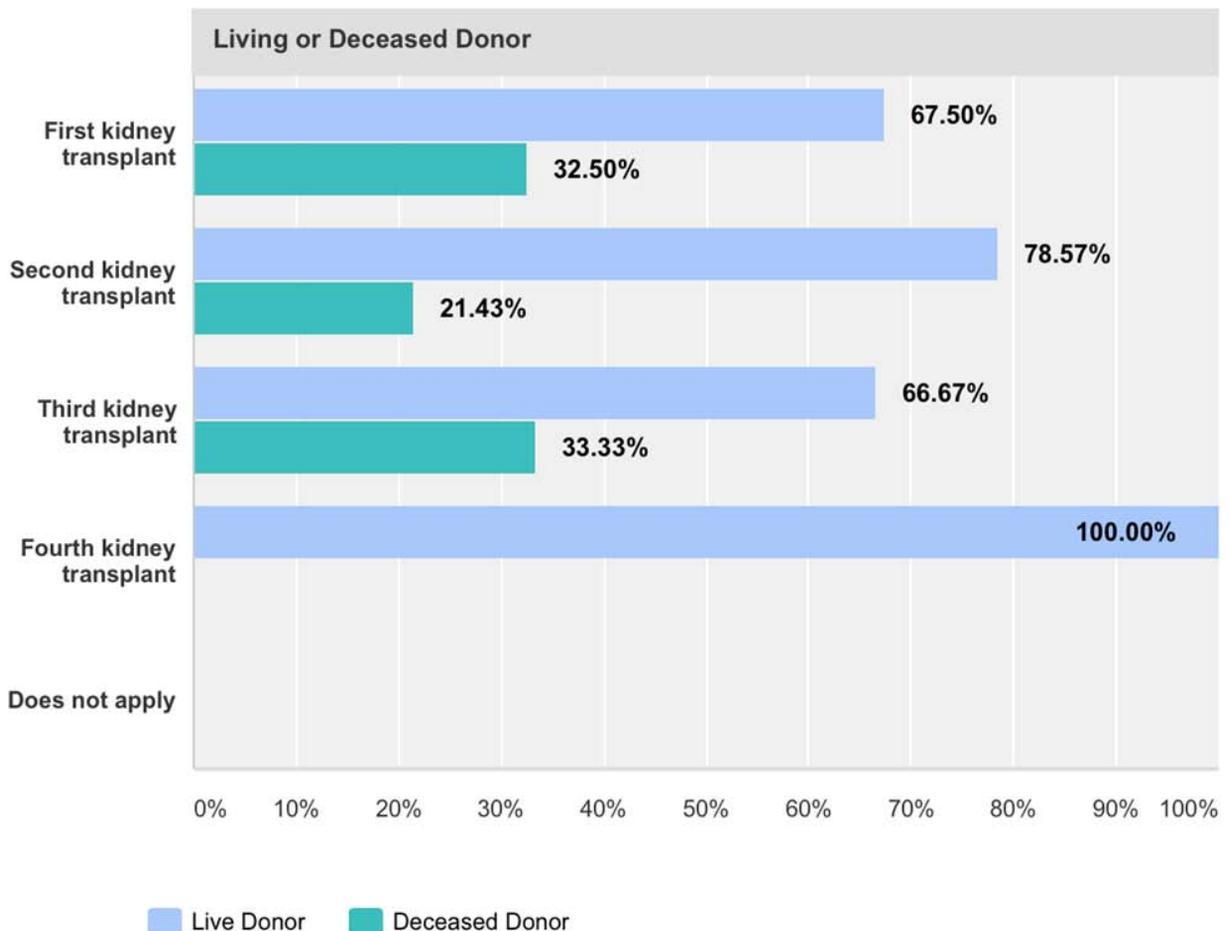
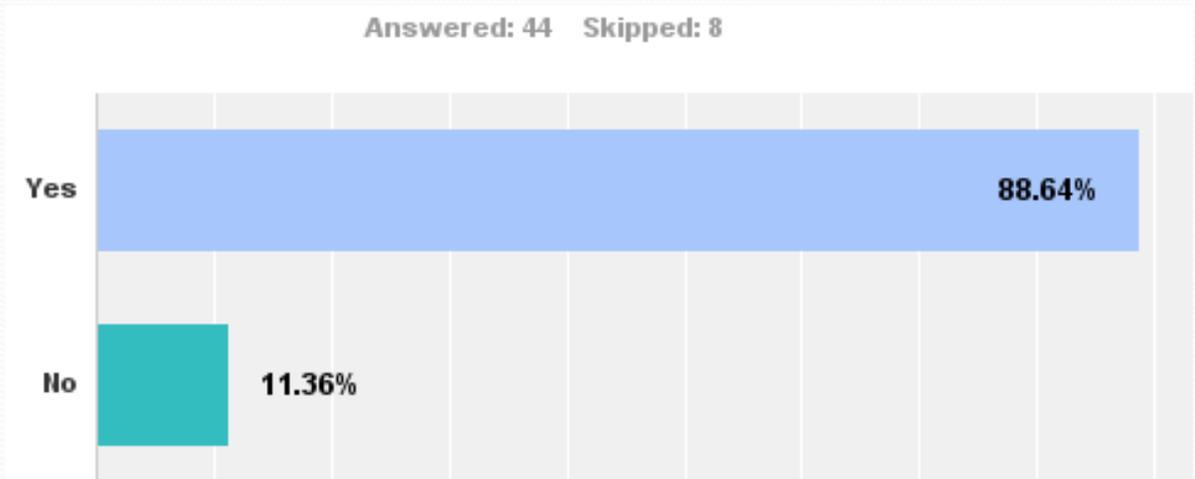
“The key for me is drinking lots of liquid. Except for the pain when swallowing, the liquid usually helps.”

“Dry things like bread are very hard to swallow. Certain shapes of certain pills are hard to get done. I wake up a lot with a bad tickle in my throat.”

“Difficulty swallowing pills... helps to drink water, repeated swallowing, regurgitating pills back up and trying again. Coughing and choking with food or liquid.... helps to stay calm and focus on breathing and regurgitate the food back up. Sensation of food/foreign body being stuck in throat - helps to clear throat, swallow a lot, drink water, and focus on breathing. Coughing or choking on saliva during non-mealtimeshelps to burp, regurgitate saliva back up, and focus on breathing. Panicking or forcing pills/ food to go down makes matters worse. To eat or drink too quickly makes matters worse. I almost always end up regurgitating whatever chokes me back up”

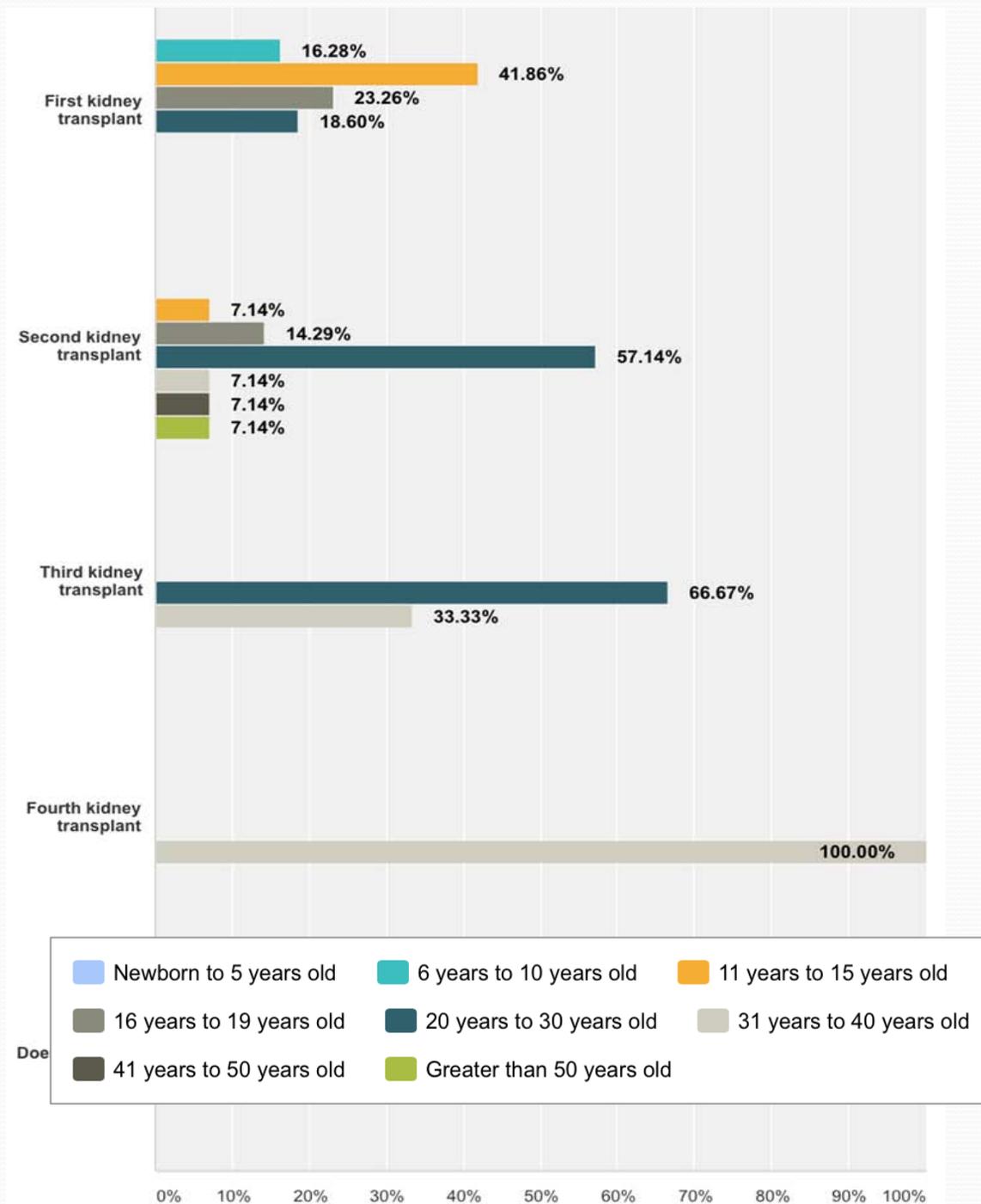
Kidney Health (Transplant) – Survey 2 (Q37&38)

Have you received a kidney transplant?



Kidney Health (Transplant) – Survey 2 (Q38)

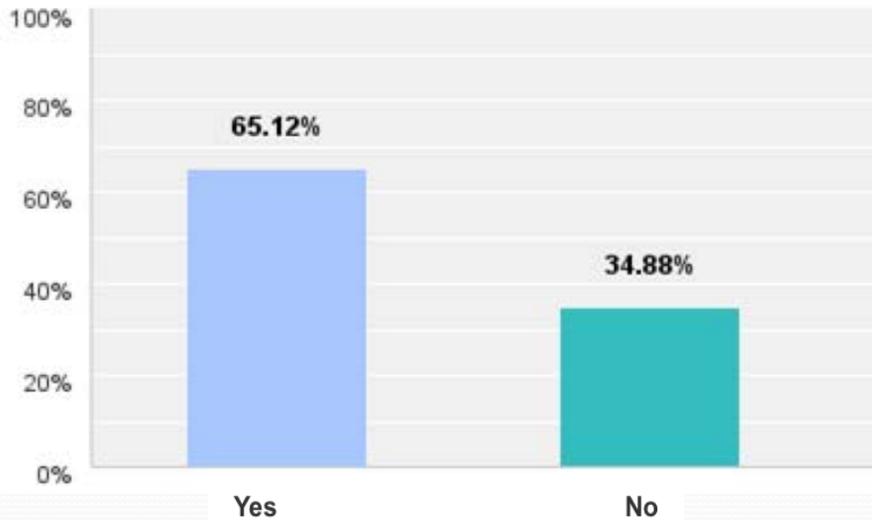
Indicate how many kidney transplants you have received and your age at time of transplant.



Kidney Health (Dialysis) – Survey 2 (Q39&40)

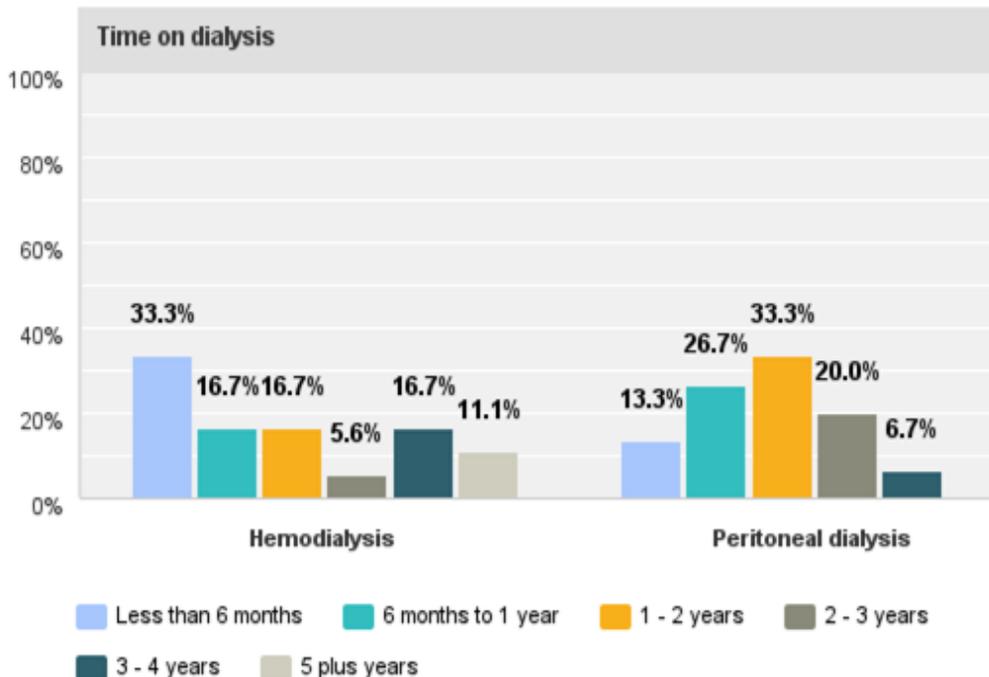
Were you ever on dialysis?

Answered: 43 Skipped: 9



If you were on dialysis, what type and for how long?

Answered: 28 Skipped: 24

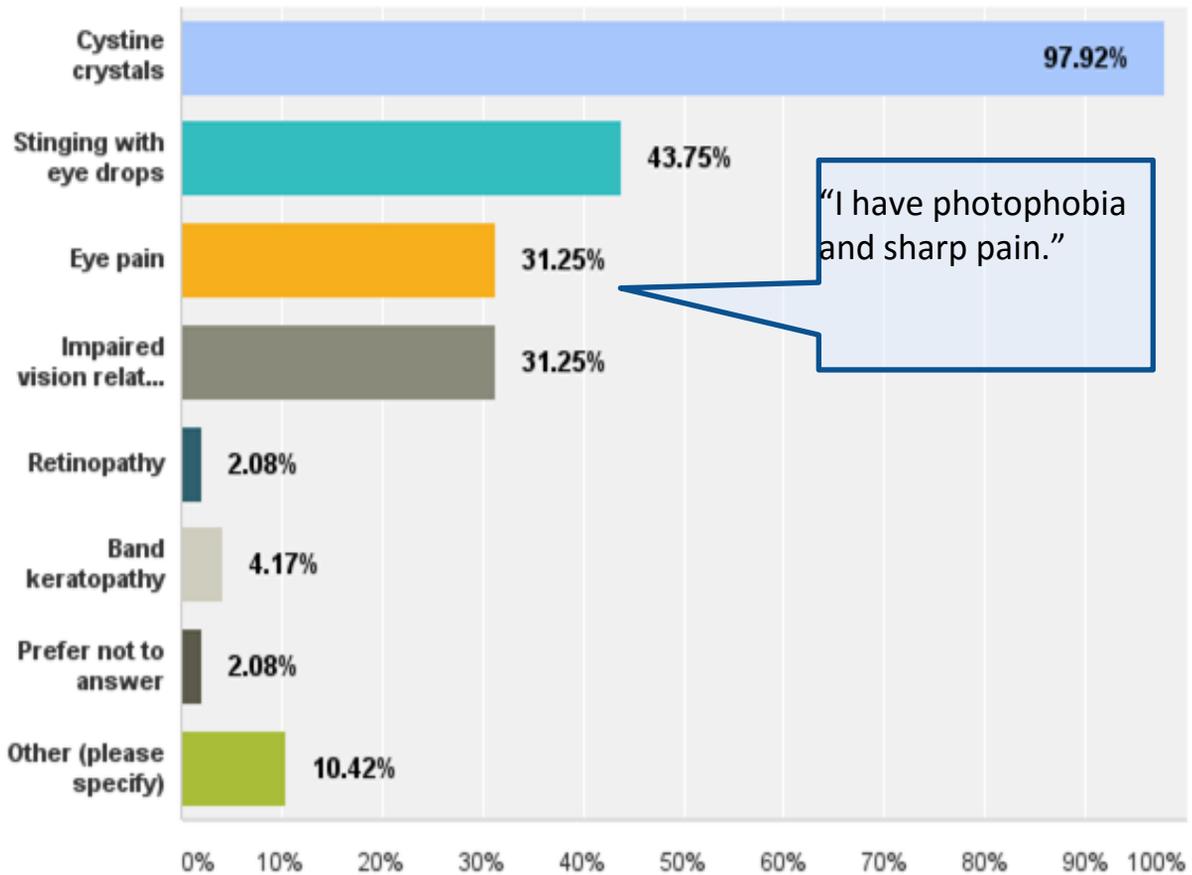


Eye Health – Survey 2 (Q17)

Do you have any of the following eye conditions?

“My eyes are crystal free. But, they always see a couple because crystals are always being made.”

Answered: 48 Skipped: 4



“I have photophobia and sharp pain.”

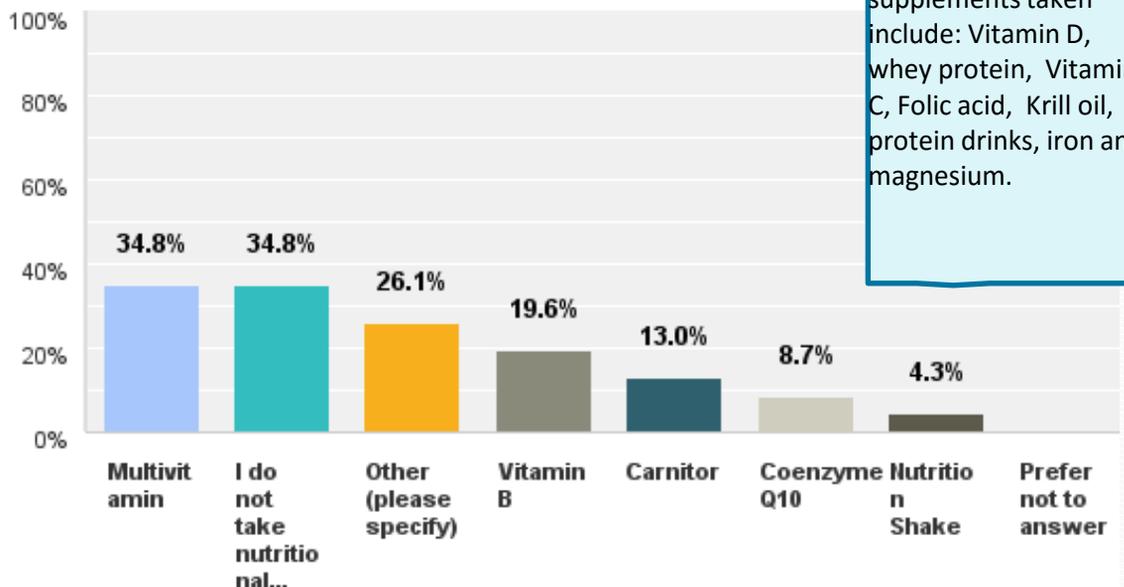
Nutritional Health – Survey 2 (Q35&36)

Answered: 46 Skipped: 6

	Disagree	Neutral	Agree	Not Applicable
I have difficulty maintaining a healthy weight	50.0% 23	28.3% 13	17.4% 8	4.3% 2
I know how to add more calories to my meals	6.5% 3	15.2% 7	69.6% 32	8.7% 4
I am confident that I get enough calories during the day, even at work or school.	15.2% 7	13.0% 6	69.6% 32	2.2% 1
I have to be reminded to eat snacks or meals.	76.1% 35	10.9% 5	10.9% 5	2.2% 1
I am frustrated because no matter what I do, I can't maintain my weight.	71.7% 33	10.9% 5	10.9% 5	6.5% 3

Are you taking any of the following nutritional supplements?

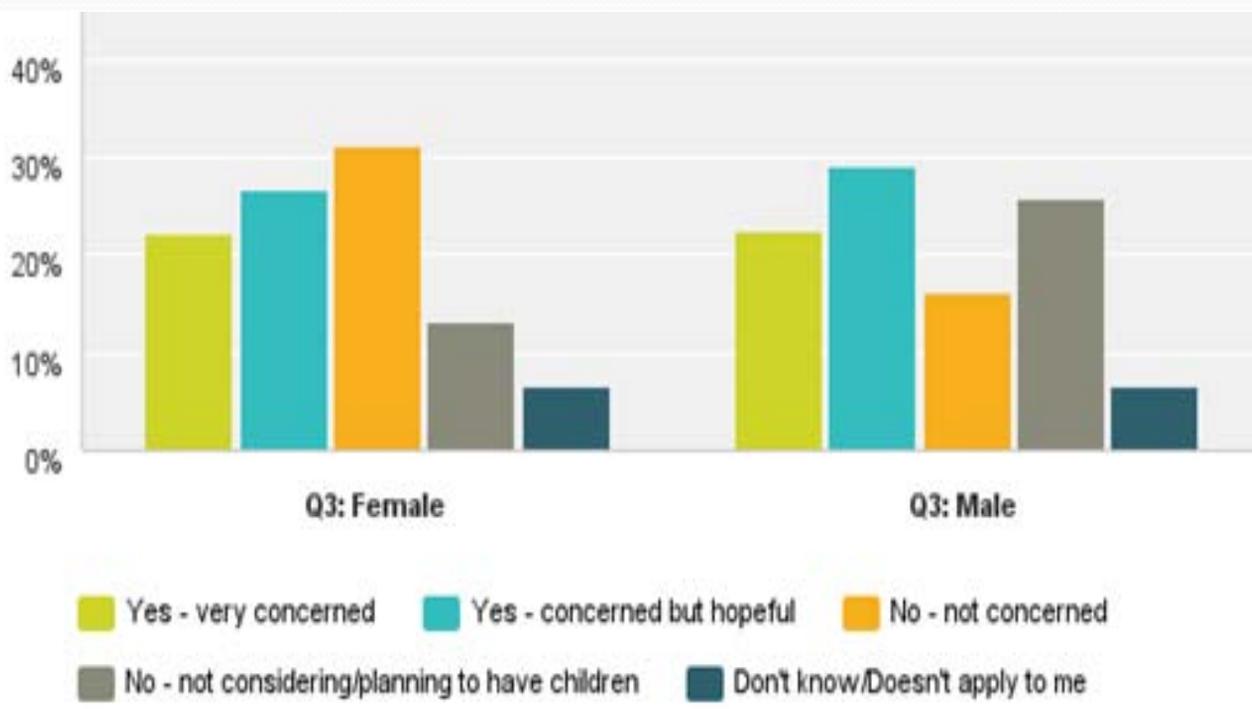
Answered: 46 Skipped: 6



Other nutritional supplements taken include: Vitamin D, whey protein, Vitamin C, Folic acid, Krill oil, protein drinks, iron and magnesium.

Reproductive Health – Survey 1 (Q18)

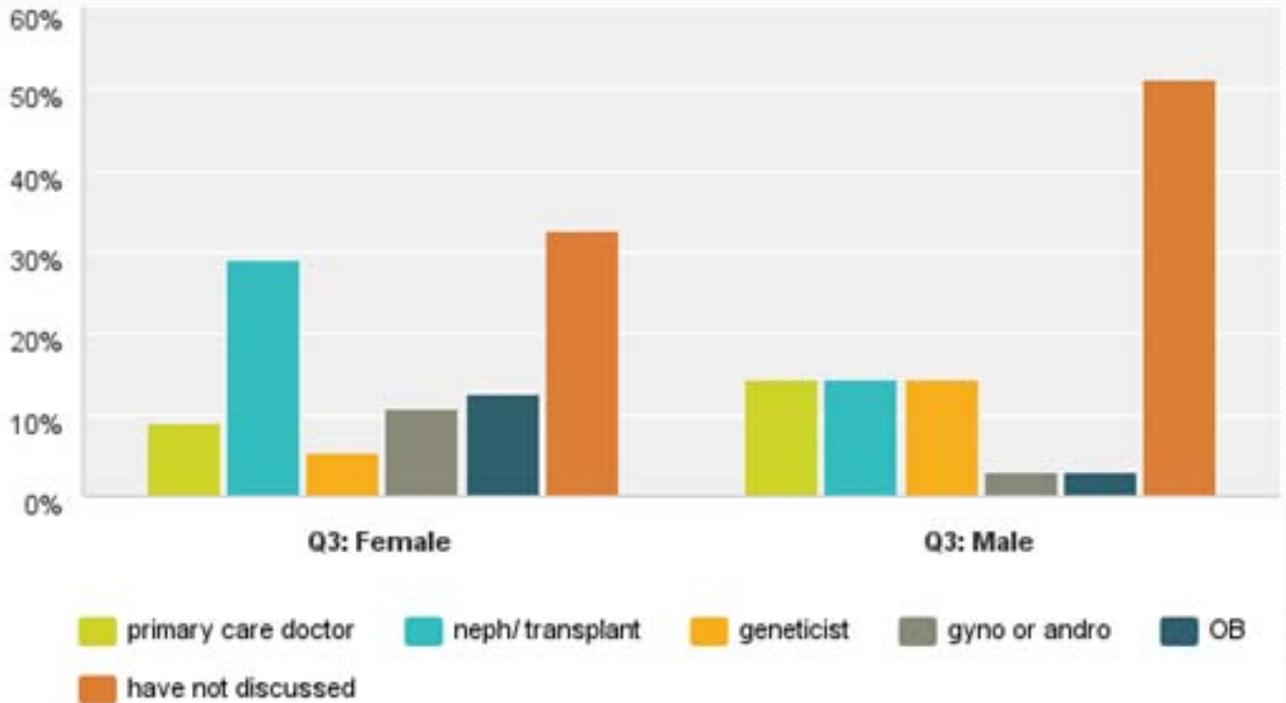
Are you concerned about your ability to have children (biological or adopted) in the future?



- A significant percentage of adults with cystinosis want to have children. Interested women want to maintain good health throughout the pregnancy (if pursuing biological children) and in the years following the delivery or adoption of a child.
- In the case of adoption, typically both men and women need to pass a health screening demonstrating that they will be able to care for a child.
- Men with cystinosis also want to have children. Many believed that this would be impossible. But with the development of assisted reproductive technology and in vitro fertilization, as well as the established system of adoption, the number of fathers with cystinosis is increasing. Unfortunately these options often come with high out-of-pocket cost and are not available to everyone.

Reproductive Health – Survey 1 (Q19)

Have you discussed reproductive health, including fertility (men) and childbearing (women) with your doctor(s)?



	primary care doctor	neph/transplant	geneticist	gyno or andro	OB	have not discussed	Total	
Q3: Female	9.09% 5	29.09% 16	5.45% 3	10.91% 6	12.73% 7	32.73% 18	80.88% 55	
Q3: Male	14.29% 5	14.29% 5	14.29% 5	2.86% 1	2.86% 1	51.43% 18	51.47% 35	
Total Respondents	10	21	8	7	8	36	68	
	Other (please describe who you have spoken to or seen)						Total	
Q3: Female							3	3
Q3: Male							4	4

Medical Concerns – In Their Own Words

“Muscle wasting is the most difficult thing in my life.”

“My lung issues are getting worse. I am often short of breath with minimal activity.”

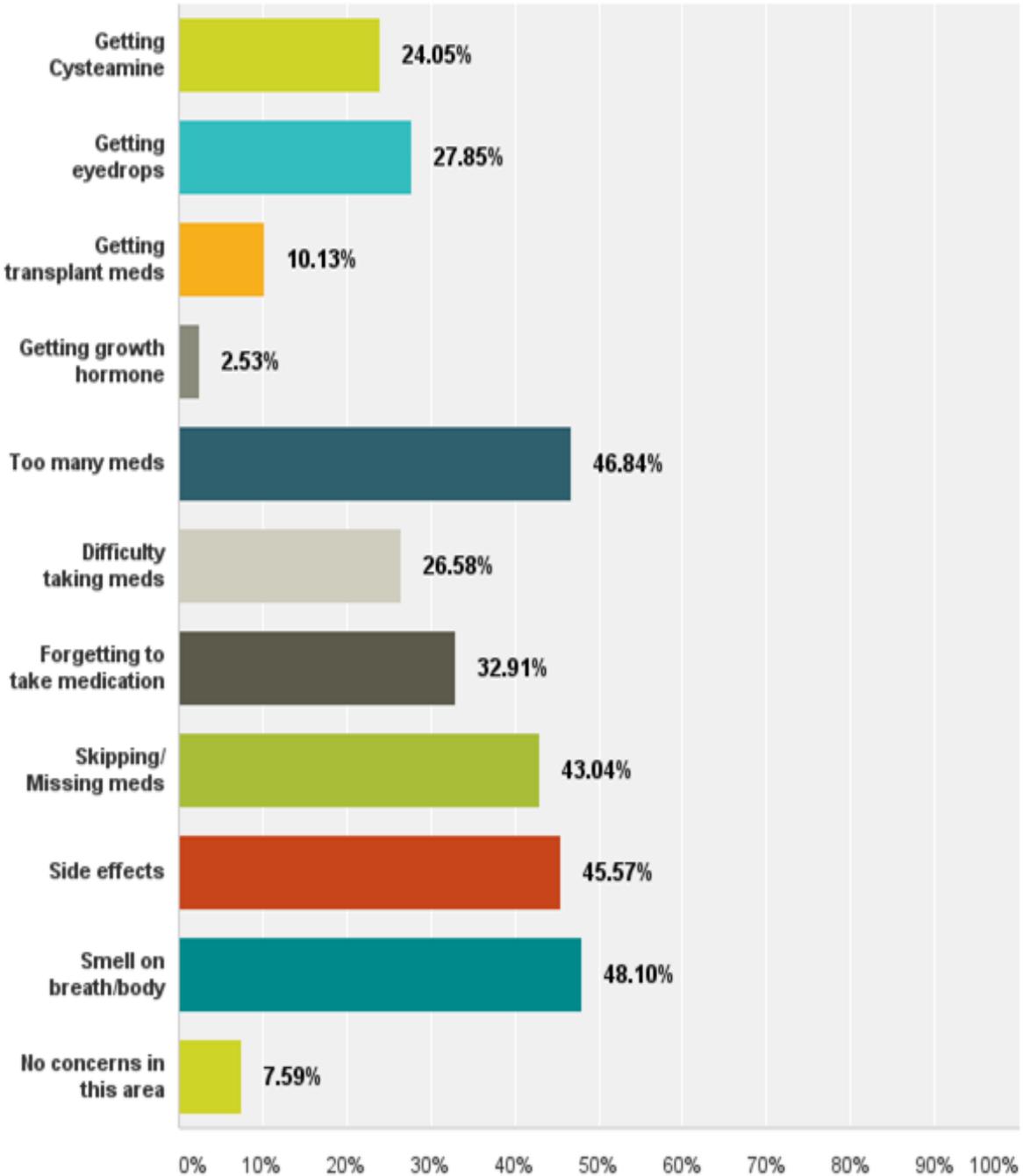
“I have difficulty swallowing pills. Even with lots of water and repeated swallowing, I regurgitate the pills back up and try again. I cough and choke with food and liquid. I feel the sensation of food stuck in my throat. I cough and choke on saliva. I try not to panic.”

“I live every day fearing my future due to transplanted kidney. I pray everything goes great for years on end. My doctor did state I will need another transplant in my future and that scares me at times.”

“Progression of disease, anemia, and low levels of energy, nausea, headaches, higher frequency of illness and metabolic specialist’s lack of help are my biggest concerns.”

Treatment Concerns – Survey 1 (Q15)

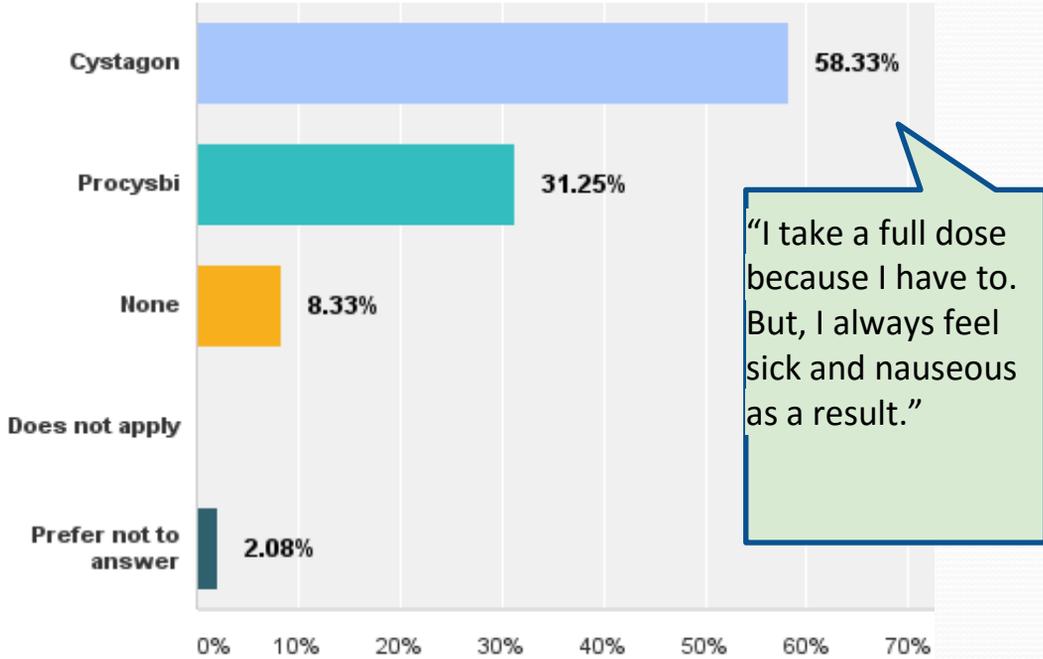
Answered: 79 Skipped: 19



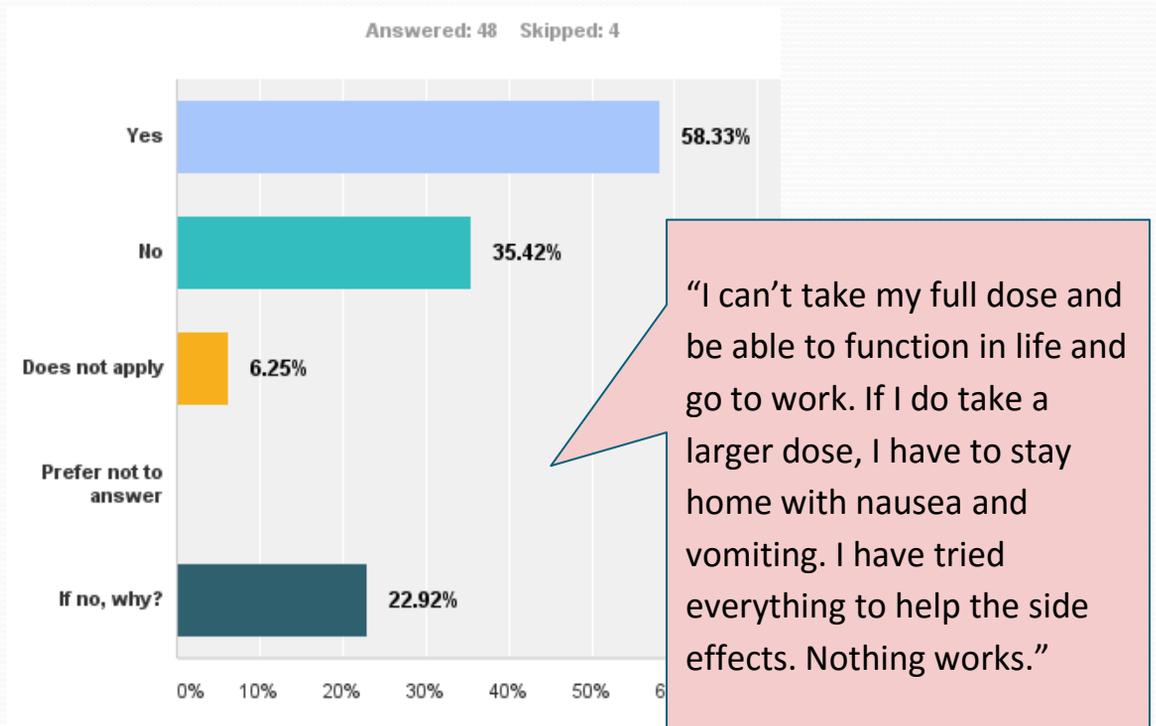
Cysteamine Treatment – Survey 2 (Q12&14)

Answered: 48 Skipped: 4

Form being taken



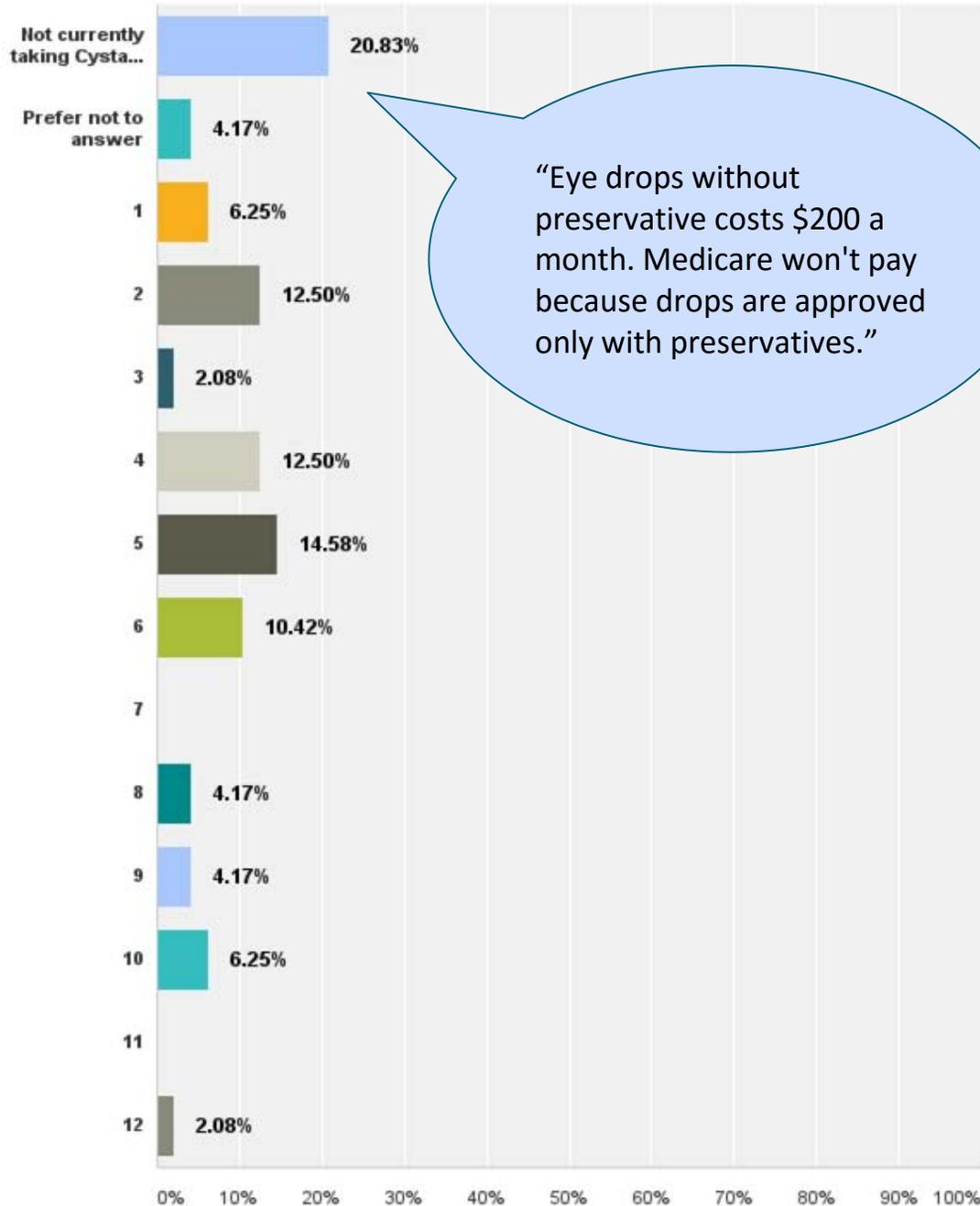
Ability to tolerate



Eye Drops – Survey 2 (Q16)

How many times per day do you use Cystaran / cysteamine eye drops?

Answered: 48 Skipped: 4



Treatment Concerns – In Their Own Words

“I have been compliant with immunosuppression for over 30 years. If I could tolerate full dose cystine-depleting therapy, I would be taking it. I don't like to hear of patients being accused of noncompliance when the issue is intolerance.”

“There is much to be improved on. I was hoping Procysbi® would be much easier to tolerate. I only take it twice a day, but must take 12 pills twice a day. This doesn't count all my anti-rejection medications. Total: 17 pills twice a day.”

“I haven't taken Cystagon® in 8 years because it made me feel so sick I couldn't get out of bed. Since stopping Cystagon® I've gotten thyroid problems, gained weight, been diagnosed with cancer, IBS, and pre-diabetes. I also tried getting back on eye drops again at the beginning of the year. They hurt my eyes so bad that I had to keep my eyes closed for about 6 hours every evening. My eyes felt like someone was putting cigarettes out in my eyeballs.”

Treatment Concerns – In Their Own Words

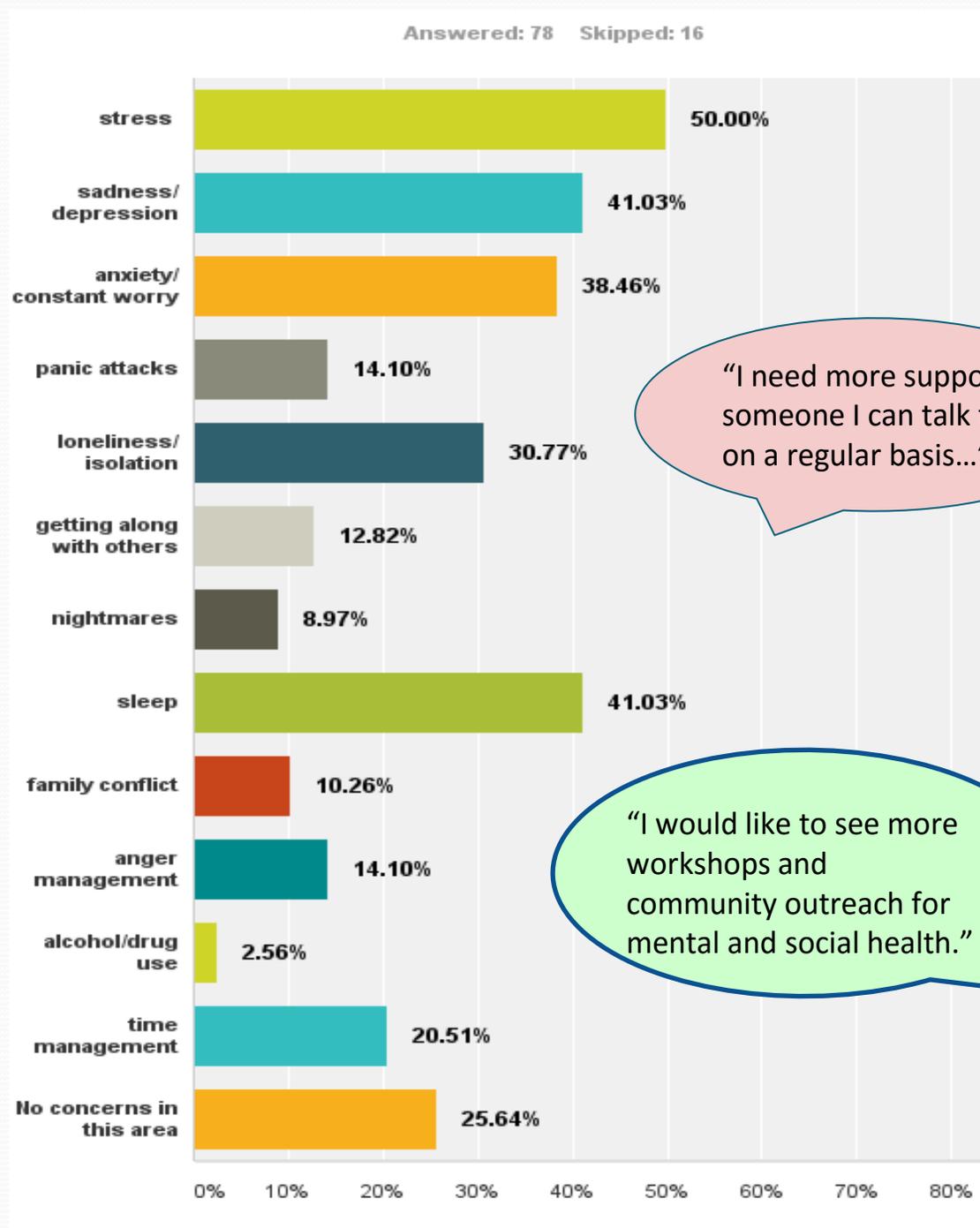
“Cystinosis patients and families are grateful for these therapies. We appreciate all of the hard work and dedication that went into the research and drug approval process. Many of us realize that there are rare diseases for which there is *no* treatment.”

“Odorless cysteamine would be great.”

“I try everything to try to tolerate more Cystagon®. The side effects prevent me from being able to work. I switched to Procysbi®. That didn't help. Because I can't tolerate my full dose, the doctors label me “noncompliant.” I wish they would understand intolerance vs. noncompliance.”

“I do not tolerate full dose Cystagon®, I never have. I have required I.V. hydration due to nausea, severe diarrhea, elevated creatinine in my transplanted kidney, and inability to sufficiently drink when I attempt to take the full dose. I have tried every suggestion given to me. I have tried nausea medications, which help, but never allow me to take a full adult dose. I have tried Procysbi®. The nausea was profound due to the longer half-life of the drug. I could not drink enough to stay hydrated.

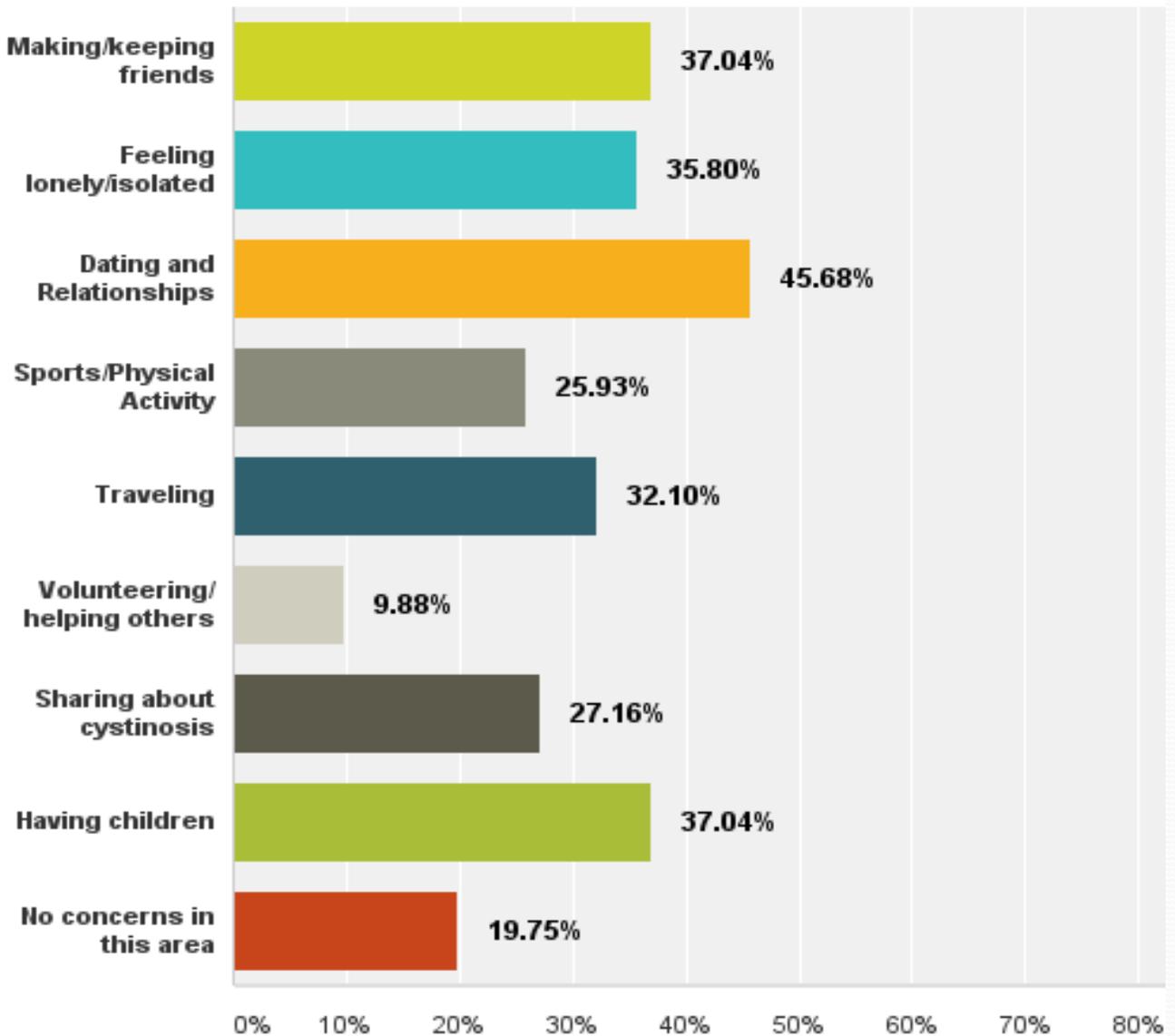
Emotional/Mental Health Concerns – Survey 1



Stress, sadness and depression, anxiety, worry, and inadequate sleep are the most powerful emotional/mental health concerns for adults with cystinosis.

Social Concerns – Survey 1

Answered: 81 Skipped: 13

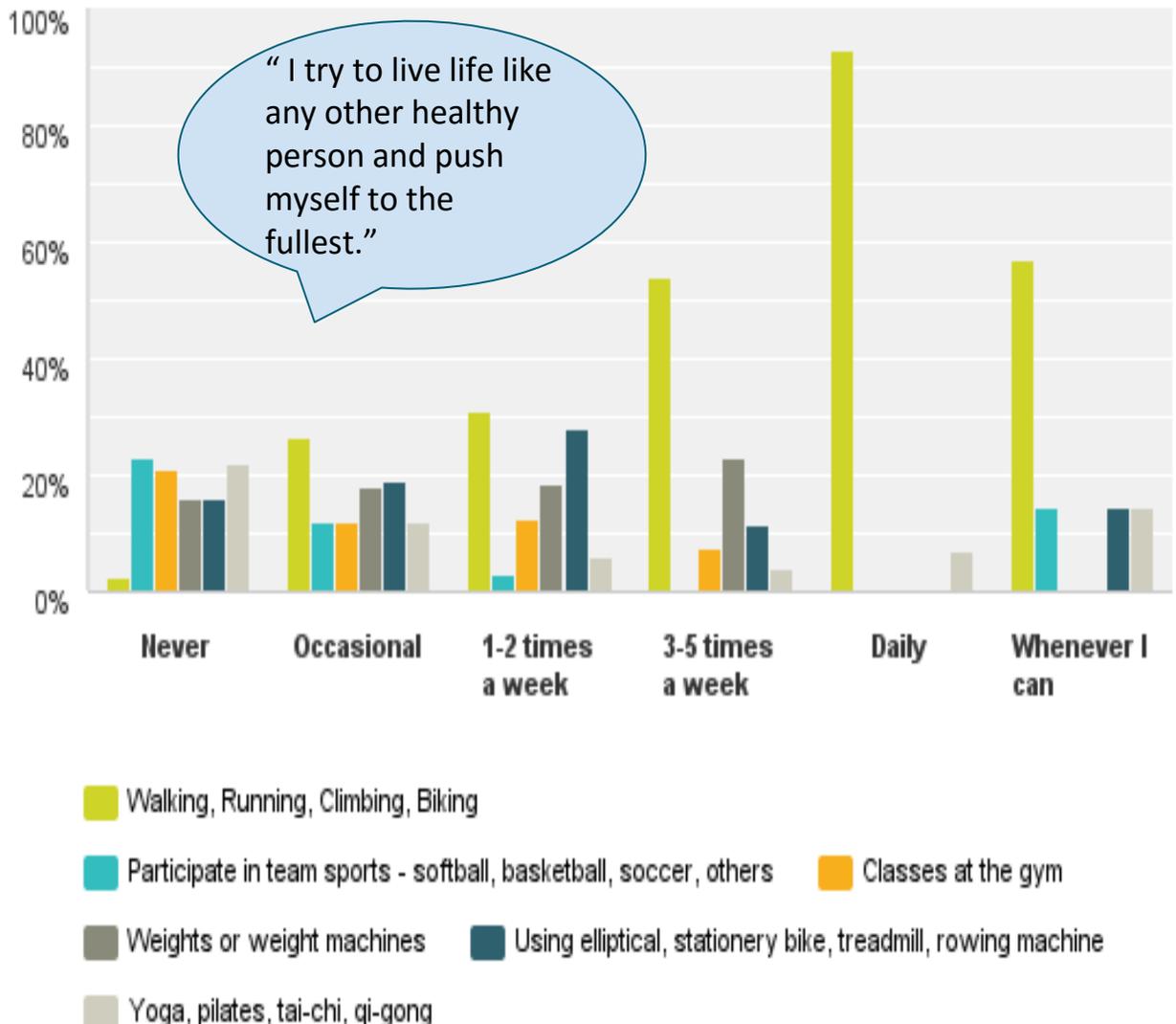


Dating and relationships, starting families, and friendships are high priorities for adults with cystinosis. Nearly 36% feel lonely and isolated.

Exercise & Fitness – Survey 1 (Q24)

What type of exercise do you do, and how often?

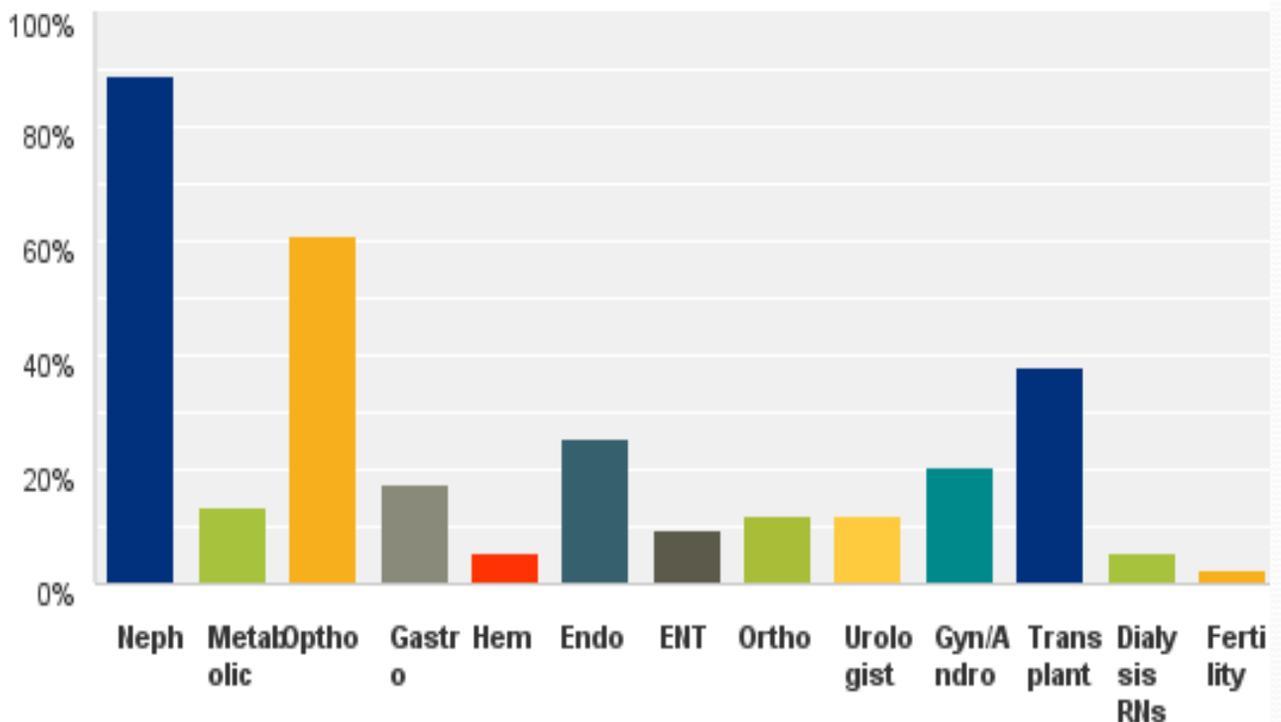
Answered: 76 Skipped: 22



Healthcare Providers – Survey 1 (Q25)

What medical specialists do you currently see?

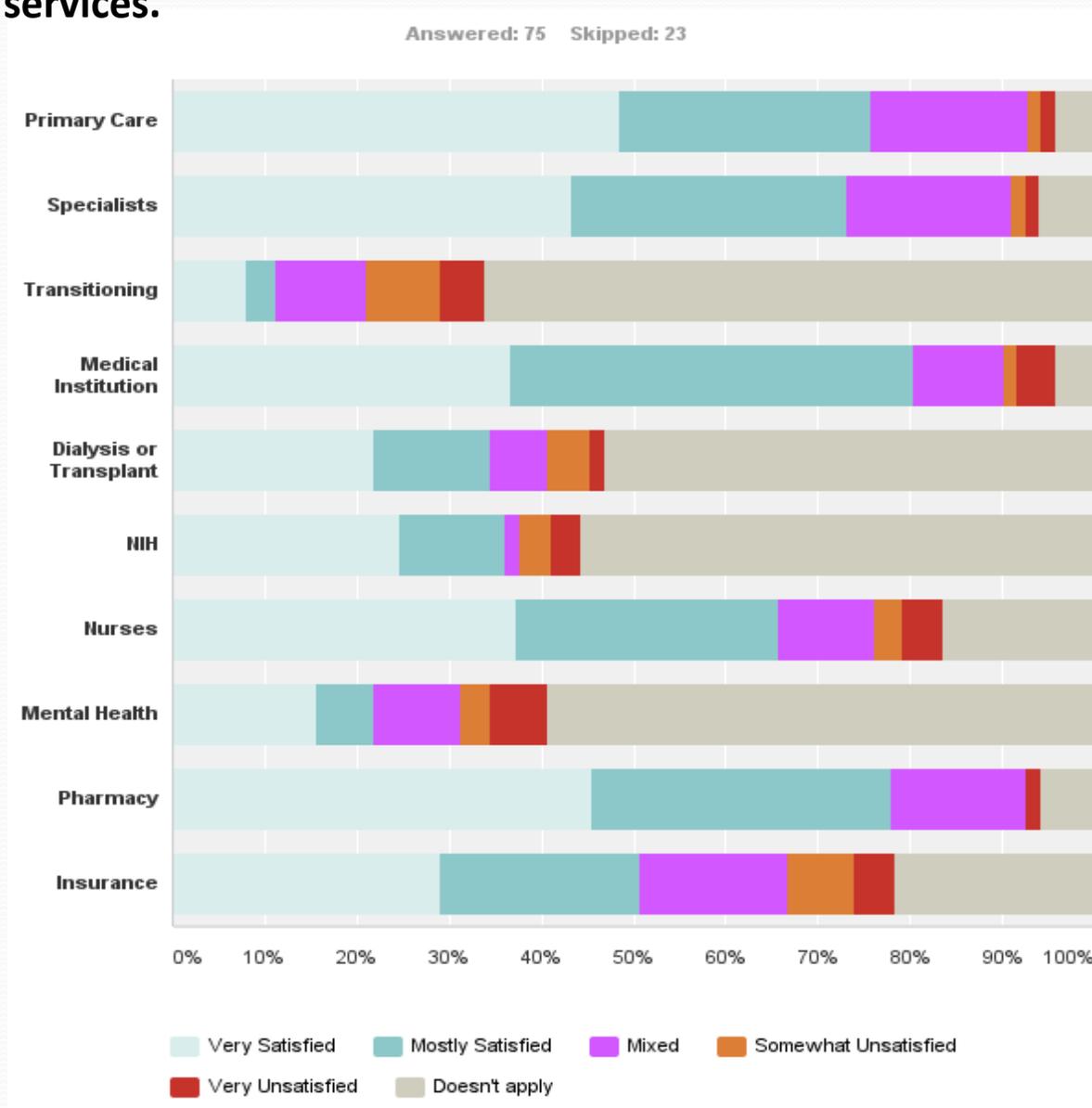
Answered: 74 Skipped: 24



Adults with cystinosis often require medical care from multiple subspecialists. Some patients aren't seeing all of the subspecialists that may be necessary. Taking time off of work to see all of these caregivers can be problematic.

Satisfaction with Care – Survey 1 (Q27)

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



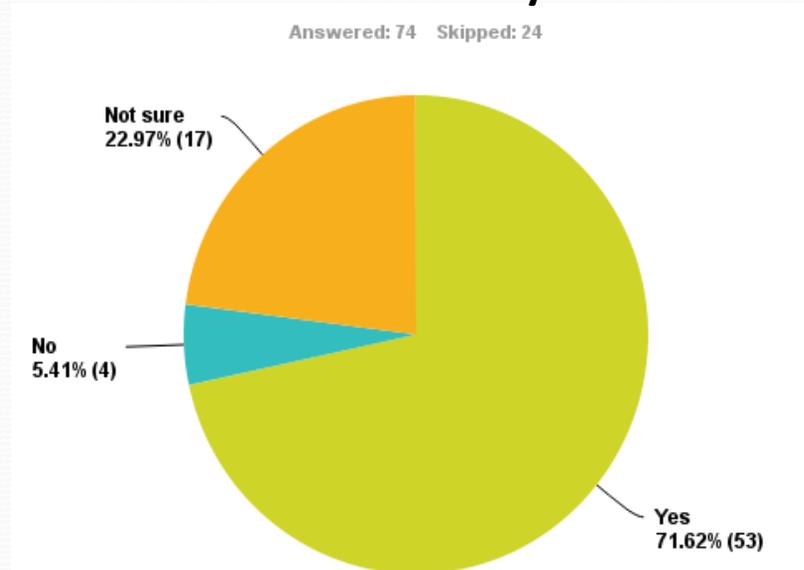
Although most adults expressed satisfaction with care, many expressed concern in finding medical teams with experience in caring for adults with cystinosis.

Participation in Research – Survey 1 (Q31&32)

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



Are you interested in being a participant in a research study about cystinosis or treatments for cystinosis?



68% of adults with cystinosis have been participants in research in the past. 72% would be willing to be participate in future research studies. There is a strong interest in research data.

Participation in Research – In Their Own Words

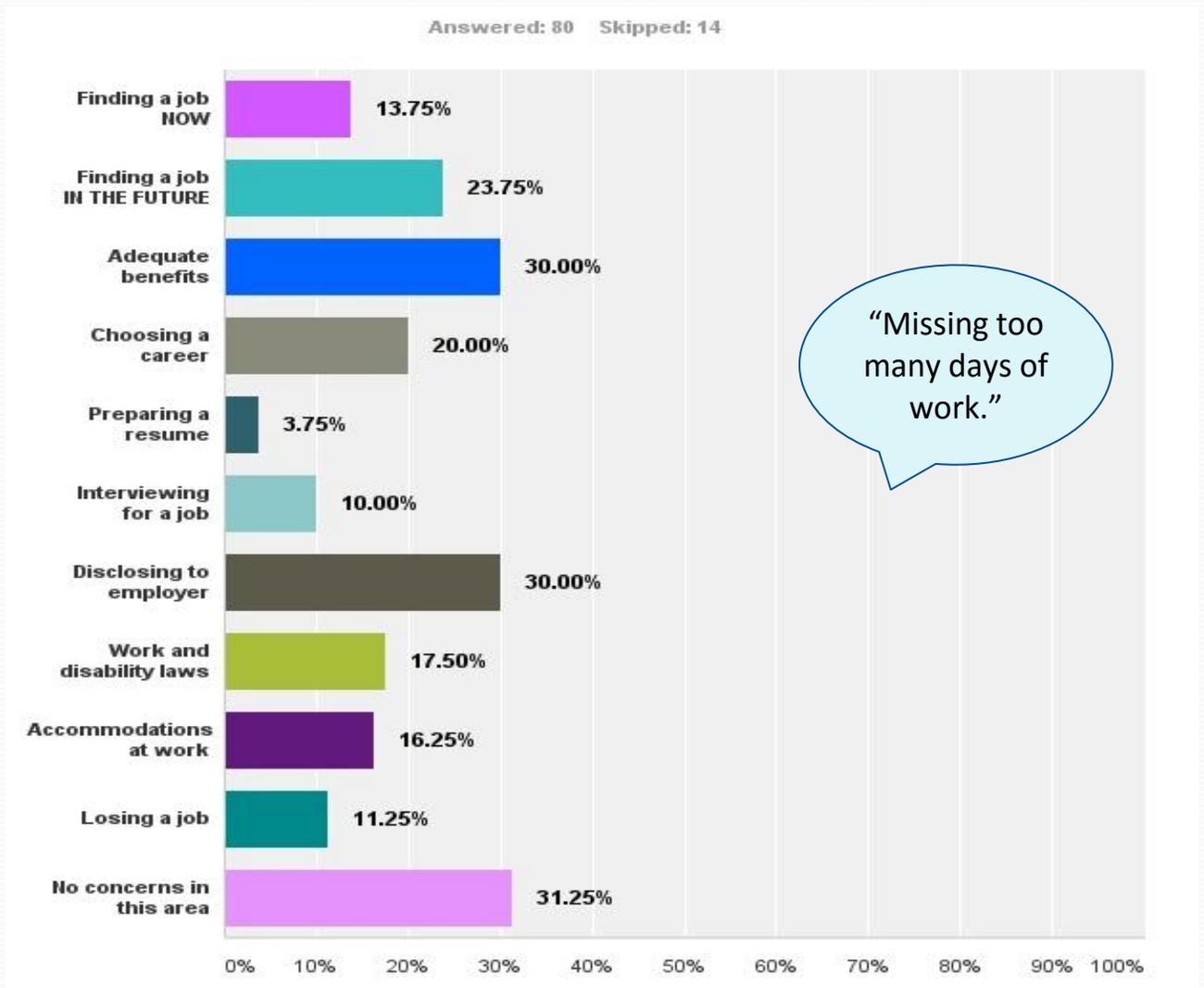
“I would enjoy being part of a study to not only learn more, but to help others with what I suffer with everyday .”

“I would like to see more research that helps adults in the future.”

“More research about life expectancy and fertility.”

“Stem cell treatments need more funding. The people who are researching or testing have a pool of people with cystinosis to choose from, and most people will do anything possible to get closer to a cure. There should also be some sort of fund for preserving/studying/testing organs of deceased people with cystinosis.”

Employment Concerns – Survey 1



Many of these concerns – such as finding a job , choosing a career, or getting adequate benefits – are similar to their peers without cystinosis. 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.

Employment Concerns – In Their Own Words

“Nobody will employ me due to illness.”

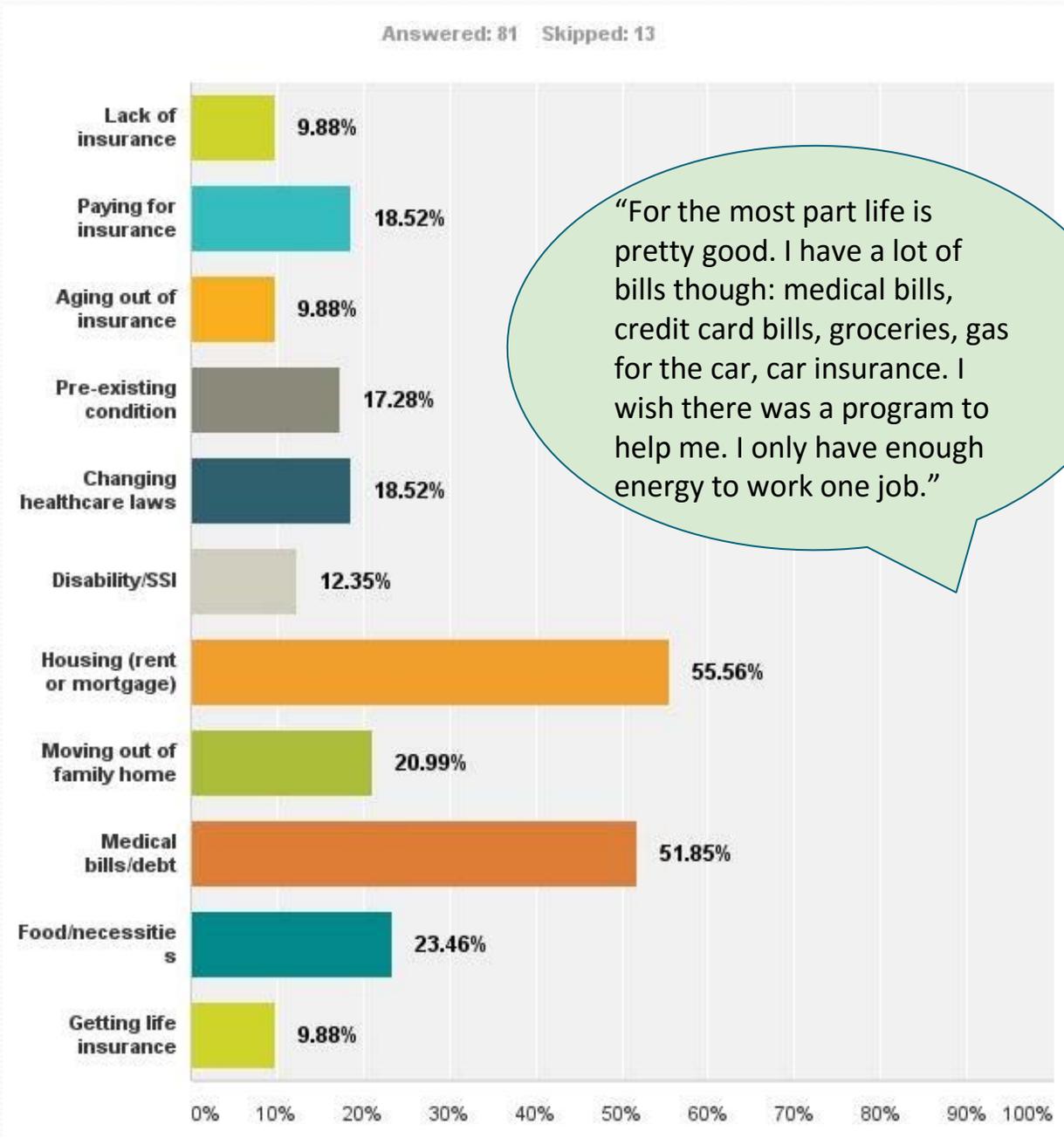
“My weak voice makes it difficult to work, to support myself, and to communicate with others.”

“I am incredibly fortunate to have worked for an employer that has a good benefit and disability package. Otherwise, I would be without food, shelter, medications.”

“Dealing with a co-worker's comments, if they unknowingly mock my side effects of cystinosis or meds.”

“Unable to work due to disability. I am incredibly fortunate for my employer's disability benefits. I get a lot of physical support from one of my parents. I worry about what I'll do if they are no longer able to help me, or if I am no longer able to help them as they get older.”

Financial Concerns – Survey 1



Adult concerns likely reflecting the employment challenges noted earlier. Adults with cystinosis are worried about meeting the costs of housing, and managing medical debt.

Financial Concerns – In Their Own Words

“Traveling to and from doctors, and eating costs.... losing a whole day’s pay because it takes 2 hours to get to my doctors and it is an all-day appointment. Insurance co-pay policies are constantly changing. Sometimes insurance covers doctor visits, meds, and labs and sometimes they don’t. Some of the cystinosis conferences are in hotels; the nightly rate is way too expensive.”

“I’m on disability and can’t afford to live on my own because I don’t make enough money.”

“Some of us are healthy enough to work, but if we work too much we won’t have our medical expenses, doctors appointments, and transplant medications covered. Some days my eyes hurt and I can’t work or the meds make me sick so I work a little slower. It’s important to find a job that works with us.”

“I have to use my credit card to pay for my hospital bills, copays, and medications . I don’t have a whole lot of money to pay my living expenses after I pay my medical bills. I only have enough energy to work one job.”

ACE Initiative Recommendations

February 2016

The following pages include recommendations based on survey findings, suggestions from patients, clinicians, and researchers from throughout the cystinosis community. Some are more easily accomplished than others, and most require funding, effort, and dedication. This is our carefully thought-out “wish list” to improve the lives of adults with cystinosis.

- Treatment
- Research
- Advocacy
- Education
- Support

For Special Consideration – Treatment

- Side effects of oral cysteamine (Cystagon® and Procysbi®), such as nausea, vomiting, and intense lethargy, prevent many adults with cystinosis from tolerating their prescribed and optimal doses. Such side effects impact daily functioning, employment, and mental health.
- Likewise, many are unable to tolerate the Cystaran® eye drops due to the preservative and experience severe pain and stinging.
- The intolerance of these medications, and thus the inability to take optimal doses of cystine-depleting medication despite best efforts, results in accumulation of cystine crystals and subsequent organ damage.
- Our understanding of adherence must include all the factors that influence how patients are able to access and utilize long-term therapies: patient-related, disease-related, treatment-related, provider-related, and system-related factors.⁵
- The adults who cannot tolerate their recommended dose are often labeled by the medical community as noncompliant. Many adults reported in the surveys that their doctors are attributing their health issues or symptoms to noncompliance rather than intolerance of treatment. The patients feel blamed and demeaned, labeled as difficult or unappreciative, rather than having their treatment challenges understood and reconsidered.

Recommendations: Treatment

Improved treatments are key to improving the outcomes and quality of life for adults with cystinosis.

- Research and develop improved cystine-depleting treatments that will allow adults living with cystinosis to tolerate and benefit from them, and live an improved quality of life.
 - Improve treatments for *all* physiological and psychological systems affected by cystinosis.
 - Pursue newer treatment options such as pharmacogenomic research and prodrugs research (*in progress*).
 - Continue research into improved ocular crystal therapies (gel, disc). Investigate issues related to preservatives and eye discomfort.
- Establish geographic Cystinosis Centers of Excellence to ensure access to care (*in progress*).
 - Comprehensive Cystinosis Centers of Excellence for adults should be established regionally in the US and worldwide.
- Develop an updated and specific plan of care for physiological and psychological systems affected by cystinosis.
- Add palliative care to cystinosis standards of care.

Recommendations: Research

Ongoing research is needed to improve the outcomes and quality of life for adults with cystinosis. We will continue outreach to adults with cystinosis through the ACE Initiative, and collect ongoing information about their concerns and challenges.

- Centralized biobank(s) for blood samples to allow for present and future research. This is critical for the type of long term research that our community needs.
- Centralized databank(s) and/or registries.
- Expand current genotype/phenotype studies.
- Pharmacogenomic studies to evaluate for potential variances in drug tolerability.
- Expand myopathy research. We are grateful for the work of researchers currently participating in this area. We have identified an additional group of researchers at Massachusetts General Hospital that are interested in collaborating on myopathy research.
- Expand mitochondrial research – Dr. Minnie Sarwal.
- Expand on long-term neuromuscular research (anatomical and functional) – many adult patients are commenting on long term memory issues. (Current cohort study in Paris, P. Cochat)

Recommendations: Research

- We need a research driven position on the use of mitochondrial cocktails. Some physicians are prescribing them, others are waiting for published data. Some patients are choosing to take these medications on their own.
- Further clinical investigation into restrictive lung disease/pulmonary issues. Invite a dedicated physical therapist that specializes in pulmonary rehabilitation to join the cystinosis community. Addition of a dedicated speech therapist would also be welcome.
- Yearly Cure Cystinosis International Registry (CCIR) updates and presentation of data to participating patients.
- Retrospective clinical studies and publication on morbidity and mortality from a single, comprehensive database.
- Dr. Matthew Sampson a pediatric nephrologist and kidney genomics researcher at the University of Michigan, is very interested in collaborating on genetic projects pertaining to cystinosis. Sampson Lab - https://sites.google.com/a/umich.edu/sampson_lab/

Recommendations: Education

Cystinosis patients need updated and accurate information to support their decision-making.

- Interactive, global web portal with information specific to the needs of adults with cystinosis, such as employment, insurance, health and mental health support, social, etc.
- Education material written in terms patients can understand to help them educate adult primary care and adult specialist physicians who may not have encountered a patient with cystinosis previously.
- Access to advocacy group conferences, meetings, and town halls through video streaming for those who cannot afford to attend. We recognize that this is an expensive endeavor, but timely distribution of information and audience participation is strongly desired by patients.

Recommendations: Support

Adults with cystinosis need ongoing social support and connection to services and resources. The psychological and social impact of cystinosis, including depression, anxiety, and the use of alcohol and drugs, must not be ignored while we seek to address the medical impact of cystinosis.

- Adult cystinosis patients are already on social media. They often learn that a friend or peer is not doing well, or passed away via this technology.
- Web portal with information specific to the needs of adults with cystinosis, with attention to overall well-being and mental health.
- Creation of a peer-to-peer mentoring program
- Creation of a reading list for support and inspiration for adults with cystinosis
- Ongoing global outreach to adults with cystinosis through the ACE Initiative, advocacy groups, rare disease support forums, and Facebook.

Recommendations: Support & Education

The family conferences, meetings, and town halls held by advocacy groups and corporate entities are vital sources of support and education for adults with cystinosis. These gatherings need to be accessible to all and include a focus on issues relevant to adults with cystinosis.

- **Technology:**
 - Content (powerpoint or video) available on the web
 - Live video streaming during events
 - For adults with quiet / weak voices, encourage participation by submitting written comments or questions
 - Suggest the use of a text-to-speech app similar to “speak it” app.
- **Participation**
 - Encourage in-person or online attendance by adults who are not currently connected with the cystinosis community.

Recommendations: Support & Education

Conference Session Ideas

- Raise awareness of adult specific physical and psychosocial issues at conferences. Some adults are hesitant to speak about disease complications in the presence of new families just learning about cystinosis.
- Breakout sessions led by both individuals with cystinosis and adult clinicians who have worked with cystinosis patients. A clinician could share what he/she addresses with his/her patients and what patients should expect from a competent local physician.
- Recruit new researchers and invite global researchers interested in adult issues to the conferences. New perspectives should be encouraged.
 - Sessions for parents of adults living with cystinosis
 - Gender-specific breakout sessions to address reproductive and sexual health.
 - Combine social events with personal empowerment activities.

Recommendations: Advocacy

- Respondents frequently perceived a lack of cooperation and collaboration, and reported a desire for a unified advocacy group.
- Creation of a global alliance of cystinosis advocacy groups to support equitable access to treatment and information, and to foster improved cooperation and coordination between advocacy groups.
- Adult advisory committee or representative on the Board of Directors of advocacy organizations.
- Interactive, global web portal with information specific to the needs to adults with cystinosis regarding areas for self-advocacy such as employment, insurance, health and health support, and more.

“Time is of the essence, there is still a lack of funding and collaboration in cystinosis. I wish the three cystinosis group would become one. Let’s bring the groups together to really marry development at the research bench and try to bring those findings to patients as fast as we can. Faster than we've ever done before.” – survey respondent

Contributors

The Adult Care Excellence Initiative (ACE) working group was formed in September 2015 with the goal of empowering adults with cystinosis, to give voice to their specific needs and concerns, and develop priorities for improved treatments, research, education, support and advocacy for adults living with cystinosis. The working group consists of adults with cystinosis, family members, researchers, and clinicians dedicated to strengthening our understanding of the challenges of and resources needed for living with cystinosis in adulthood. The working group is not directly linked to any advocacy organization, research institution, or corporate entity.

Meet The ACE Initiative Working Group Members

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We welcome the addition of new members from all countries. Please contact the working group: CystinosisACE@gmail.com.

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