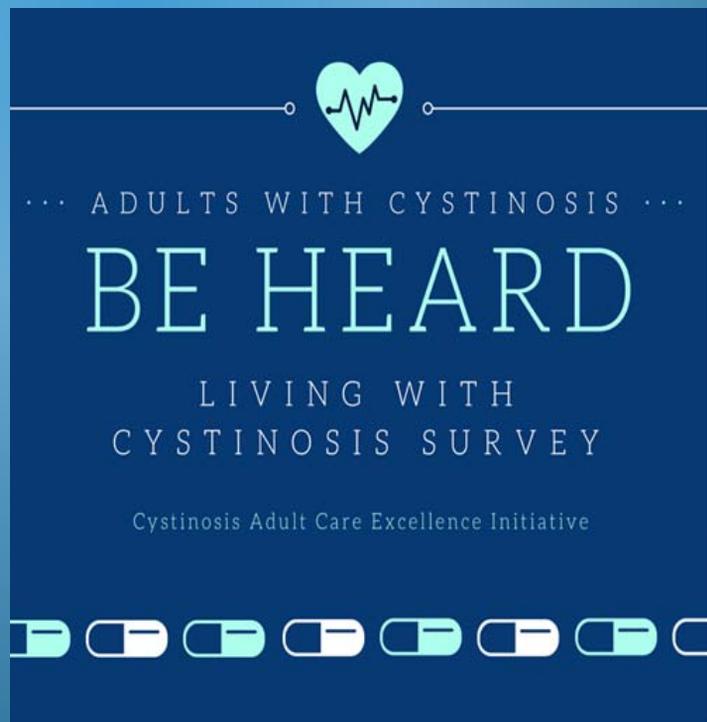


Cystinosis Adult Care Excellence (ACE) Initiative

Executive Summary

Initial Survey Results and
Recommendations
February 2016



Executive Summary

Cystinosis Adult Care Excellence Initiative

Cystinosis was seen for many years as a disease of childhood, one primarily managed by pediatric nephrologists. Lifespans were curtailed by the disease, often in adolescence. Breakthroughs in medical interventions, including cystine-depleting medications, kidney transplant, and other treatments, have allowed individuals with cystinosis to survive – and thrive – well into adulthood. We now have a growing population of adults with cystinosis. Many of the patients in our adult population selflessly served as research subjects for the drugs and protocols that are now helping newer generations of patients. For the first time ever, we are now looking at a future of relatively well children and young adults with cystinosis. In contrast, we are also seeing an increased population of older adults who face a broader range of health issues. Cystinosis can no longer be referred to as a disease of childhood; it is a chronic disease that impacts individuals across the lifespan, and which presents unique medical and psychosocial challenges for adults.

The incidence of cystinosis in the United States is 1 case per 100,000 - 200,000 live births¹. An estimated 400-500 people in the United States have cystinosis. A higher incidence of cystinosis, 1 in 26,000 live births, has been reported in the western French province of Brittany². Many children and adults with cystinosis continue to live with life-limiting effects. Their lives' trajectories are often dominated by cystinosis, with the disease and health problems influencing nearly every aspect of their lives; in many cases, cystinosis affects relationships, employment, having children, and financial stability. Successful transition from pediatric to adult care plays a key role in disease management and addressing the emerging long-term challenges associated with cystinosis. This period of transition marks a shift in disease management from the pediatric nephrologists (in many cases) and parents, onto the shoulders of the young adult with cystinosis. Independence, self management, and treatment adherence are expected and required for good clinical outcomes.

The 2015/16 **Living With Cystinosis Surveys** are the first effort to take a comprehensive look at the total impact of this disease on the adult population. These surveys successfully capture the voices of more than 146 adults living with this disease to date. They look at an array of concerns including medical care, treatment, psychological issues, social wellness, and financial burden. The survey results and recommendations presented here are the result of input of adult cystinosis patients living in several countries around the world. Over time, we hope to obtain even greater participation worldwide. Adults living with cystinosis are extremely resilient and resist being defined by cystinosis. Despite the fact that some of the older adult patients had early diagnosis and access to treatment, many are now facing multiple medical and psychosocial challenges. We are losing patients in the prime of their lives.

TREATMENT

There exists a broad-based misperception that nonadherence is synonymous with intolerance with regard to cystine-depleting therapies that cause a number of uncomfortable and at times intolerable side effects. This has added a very painful challenge for adult patients and parents. For many adults, this “Nonadherent” label is crushing, as patients are often fully compliant with immunosuppressants and other medications. Rather than continue to label these patients (both children and adults) as nonadherent, it is time to develop therapies that are better tolerated.

The top four medical concerns presented in these surveys are: kidney or transplant status, muscle wasting in smooth and skeletal muscles, deteriorating speech and swallowing, and pulmonary/breathing issues. For many years we have known that the muscle deterioration associated with cystinosis resulted in restrictive lung disease and breathing issues. Our surveys demonstrate that three of the top four concerns are connected to muscle wasting.

Previous data show that cystine-depleting therapies like Cystagon® and Procysbi® “improve the outcome and complications of cystinosis, but they do not prevent them”³. Gene therapy holds promise for the future; however, not every patient with cystinosis will be eligible for this form of therapy. There is a critical need today for research that will result in newer therapies with less disruptive side effects. Improved treatments are the key to improving the medical outcomes and quality of life for adults with cystinosis. We need a worldwide effort to develop and make available tolerable cystine-depleting therapies to as many cystinosis patients as possible, so as to allow all adults living with cystinosis to tolerate and benefit from them. We find ourselves at a moment in time that requires immediate action. We must also identify treatments for the many long-term medical issues that emerge for adults with cystinosis. Far too many adults with cystinosis are dying prematurely because of these medical issues.

Health often plays a dominant role in the lives of those with cystinosis. The adults in the community are pioneers, who with increasing age find that further health-related issues emerge. We must recognize the altruistic role they have played, taking part in formal and informal research for most of their lives. While some adults will experience few or mild complications, many are concerned that they are simply waiting for the next problem to be diagnosed. Although this is quite stressful, and the average person would be mentally exhausted and overwhelmed, adults with cystinosis have come to accept this as part of the disease.

In addition to the kidney issues and muscle deterioration concerns, adults with cystinosis have identified challenges with: high blood pressure, vision/eye pain, hypothyroidism, gastro-stomach issues, persistent headaches and nausea, memory problems, joint pain, fatigue, diabetes, dizziness, and bone problems. Issues of low libido, fertility, and pregnancy are also important concerns for the adult population, as their future plans mirror those of their friends and peers.

The issues associated with muscle deterioration are not unlike some of the issues faced by people with other neuromuscular disorders, and therefore there may be treatments currently available or research underway to address some of these concerns within other diagnoses.

We should investigate pharmacology and genomics (pharmacogenomics) and technological advancements, as well as telemedicine and complementary therapies.

To ensure access to quality care for people with cystinosis, developing Cystinosis Centers of Excellence or a National Center of Excellence that is connected to smaller Regional Centers will significantly reduce the geographical barrier to care that often precludes people with physical disabilities' financial constraints.

Finally, it is extremely important that we build on previous work and develop updated Standards of Care for cystinosis that include the many issues associated with the late-onset challenges that adults face.

RESEARCH

Future research will identify new and improved treatments. In addition to gene therapy and improved cystine-depleting treatments, there is an immediate need for research associated with the neuromuscular degenerative process. Myopathy issues affect most adults with cystinosis, and many adolescents. With few exceptions this issue will in some way impact everyone with cystinosis as they grow into adulthood. To optimize data collection for research, centralized biobanks and databases should be established and maintained.

SUPPORT, EDUCATION, & ADVOCACY

To improve the ability of adults with cystinosis to make sound medical decisions, they will need accurate information. A web portal is needed containing information specific to the needs of adults regarding medical treatment, mental health, employment, insurance, and social support.

As adults transition to a primary care physician and adult-oriented specialists, they often assume the burden of locating, and then educating, their new providers. Education materials and support are needed to further assist with this transition.

By leveraging technology we can enhance the participation of adults with cystinosis in advocacy or education conferences, and reduce the geographic or financial barriers. This can be accomplished through live streaming and webinars, social media, and text-to-speech apps for those in attendance with quiet or weak voices.

The cornerstone for cystinosis education has always come from the many conferences hosted by this community. As we look to improve care, advocacy, and the quality of life for adults living with cystinosis, it will be important to consider sessions that raise awareness for these issues, breakout sessions led by adults, gender-specific sessions on reproductive issues, and social events for adult empowerment and networking.

Finally, unifying the great work of our cystinosis advocacy groups is a high priority to ensure that we speak with one strong voice. A consolidation of the advocacy groups and/or meetings could strengthen these efforts immeasurably. The formation of an adult advisory committee to specifically represent and advocate for adults living with cystinosis is long overdue. This would send a positive message and provide a voice to this underserved group.

CLOSING THOUGHTS

There are many challenges ahead for both patients with cystinosis and the clinical teams caring for them. As advocates for all people with cystinosis, we have a responsibility to optimize the conditions for the success of adults living with cystinosis. We need to work in partnership with them to provide the best possible care. The majority of children with cystinosis are growing up to reach adulthood. These adults will require medical interventions that are currently unaddressed/no treatment exists. They need our ongoing support so that they can live, or continue to live, fulfilling and active lives. Most of the adults with cystinosis have spent a lifetime supporting research for future generations; we must now invest in them.

Respectfully submitted,

The Adult Care Excellence Initiative

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