Advancing Adult Leadership in Cystinosis Advocacy and Support

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Introduction

Cystinosis is a rare metabolic disease that causes the amino acid, cystine, to accumulate throughout the body. The disease affects every cell in the body, with particular impact on kidneys, eyes, and muscles. It is now typically diagnosed in early childhood. Thus, diagnosed patients have been aware of their illness from as early as they can remember.

Earlier diagnosis and improved treatments over the last 35 years - including kidney transplant, immunosuppression drugs, and cysteamine treatments - have extended the lifespans of individuals with cystinosis from adolescence well into adulthood.(Doyle & Werner-Lin, 2015; Nesterova & Gahl, 2013) A clinical trial of gene therapy for cystinosis is now in progress (CITE).

In the past, individuals with cystinosis often died in childhood or adolescence. There is now a generation of cystinosis patients who are surviving and thriving into adulthood, yet still face many challenges including disease progression and complications, and the impact of the disease and its treatments on their daily lives and decision-making. Advocacy for support, improved treatments, and ongoing research has until now been the task of caregivers. Adults with cystinosis are now taking an active role in advocacy for themselves and for their community.

ALAB 2018-2020 Members

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Steve Schleuder
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in memory of Laura K. McGinnis

Evolution of ALAB

The Cystinosis Research Network (CRN) was founded in 1996 by a "kitchen table" of parents/ care-givers and has since become an international force in raising awareness, fundraising, patient and family support, and research advocacy. As patient survival lengthened, CRN began to take a greater interest in the transition to adulthood and adult-oriented healthcare, publishing the first Bridges to the Future transition guide in 2011.Since then, more formal patient/family education materials, sessions at bi-annual national advocacy meetings, two "Cystinosis Teen Adventures" hosted by Global Genes, and a series of Future by Design regional town halls have been undertaken to meet the needs of this growing adult patient community.

ALAB Projects

- Cystinosis SESSIONS video conferencing– provides a face to face online platform to share experiences and educate cystinosis patients, parents, caregivers, and healthcare professionals in a knowledgeable and comfortable environment.
- Cystinosis RARE: A Journey Into the Unknown - quarterly Podcasts, hosted by individuals living with cystinosis, delivers community and provider input, highlighting topics such as mental health, cystinosis success stories, teen to adult transition, and educational system challenges.
- Cystinosis TEENS Instagram Updates – provides a private/ closed Instagram account, moderated by ALAB members, focused on challenging issues, spotlighting individuals and highlighting successes, as a safe forum for teens.

The Adult Leadership Advisory Board (ALAB) of CRN was convened in 2018 as a formal way of empowering a generation of patients who are now surviving to adulthood. The future leaders of the organization (and of the wider cystinosis community) will be a combination of patients and caregivers. ALAB leaders are appointed on a rotating basis, to nurture and support shared leadership and develop leadership skills across the community (similar to the board structure of the larger foundation). Members apply and are selected for 2 years terms. This new structure echoes what has occurred in the leadership of other organizations including the NKF, which was originally founded by caregivers and whose current CEO is a kidney transplant recipient with a familiar history of kidney disease.(NKF, 2015)

Peer support amongst those with chronic health conditions has been demonstrated to improve knowledge and coping, improve communication with healthcare providers, support better adherence to complex regimens, and to empower both individual health decision-making and advocacy efforts (Doull, O'Connor, Tugwell, Wells, & Welch, 2017) Social support and the influence of others, both in person and through social media, can have an impact on personal health behaviors and quality of life (Doyle, 205)

ALAB's intention is to nurture other young patients who have cystinosis, have a positive impact and provide guidance along the disease process with hints, tips, and companionship to reduce isolation.

ALAB's mission is "***to share our stories and strength to educate, motivate and empower the entire cystinosis community***";
The ALAB philosophy is that **those who have lived it, can teach it**.
For 2019-2020, ALAB is developing projects with the goal of using newer communication technologies to reach crucial adolescent, young adult, and adult constituents with cystinosis. Brief survey instruments have been created and will be disseminated by hyperlink and email after each project event. New projects will be developed over time as ALAB leadership rotates and new concerns arise.

Implications

ALAB aims to help teens, young adults and adults diagnosed with cystinosis, balance their lifestyle choices with the demands of managing their health condition. By connecting cystinosis patients, ALAB can encourage positive outlooks and behaviors, and improve term outcomes of those newly diagnosed.

Recipients of ALAB content can strengthen their knowledge of cystinosis (including treatment options and adherence); impact other aspects of their lives such as work, employment, and relationships; navigate the healthcare system; and build problem-solving skills. Those creating ALAB project content will develop teaching and leadership skills and motivation to maintain and strengthen their own disease management and coping. While the lifespan with cystinosis has expanded due to treatment innovations, patients must continually refresh their knowledge and make active decisions regarding their health and healthcare. They must remain advocates for research, treatment, and improvements in quality of life. In the long term, ALAB aims to help improve the health and mental health of adults with cystinosis, empowering them within their own lives and healthcare systems, and within the broader community.

ALAB members, representing this rare disease community, want cystinosis healthcare providers and researchers to recognize that young adult and adult cystinosis patients have become well-versed in their own disease, and can clearly speak about how the disease impacts their lives. Most strongly, they encourage healthcare providers to **LISTEN**. Patients with cystinosis are now active consumers of information and communicate within the cystinosis community to identify shared concerns and needs. They can and should be active members of their own healthcare teams, and can educate their physicians, nurses, nutritionists, social workers, and other specialists and mental health providers about many aspects of living with cystinosis.

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