

# Family Strategies for Living with Rare Disease: The Experience of Cystinosis

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**ABSTRACT** *Objective:* Medical advances have transformed the rare disease cystinosis from fatal in childhood to chronic and manageable well into adulthood, creating new challenges for patients, families, and providers. In response, families are adapting strategies to meet the demands of the illness and its treatments during a developmentally dense stage of life. *Method:* The study uses a classic grounded theory approach to understand the experience of adults and emerging adults living with cystinosis. Data were collected through focus groups and/or semi-structured interviews with 46 individuals (22 patients and 24 parents) recruited through national cystinosis advocacy/support organizations. Constant comparative analysis guided data collection and analysis. *Results:* In the context of much uncertainty, these patients and families described interconnected strategies to negotiate childhood and adolescence, enter into adulthood with self-care skills, and integrate normative developmental tasks with adequate management of illness. Strategies included educating, regimenting, pushing/protecting, connecting, coming to terms, and finding a fit. *Conclusions:* As both the understanding of and treatment options for rare and genetic childhood diseases expand, health care providers and multidisciplinary teams should consider regularly assessing the strategies used by families to manage illness. Families and individuals adapt as treatment protocols evolve and as children survive into adulthood, facing new, and perhaps unexpected, developmental tasks. The strategies described by participants can help frame the expectations of families across rare disease groups and suggest new avenues for resources and support.

**KEYWORDS:** cystinosis, transitioning, adherence, emerging adulthood, grounded theory

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Cystinosis is a rare lysosomal storage disorder that results in intracellular cystine accumulation in organs and tissues (Nesterova & Gahl, 2013). With a prevalence of one case per 100,000–200,000 live births, an estimated 500 nephropathic cystinosis patients are living in the United States with an addi-

tional 2,000 patients worldwide and about 15 new diagnoses made per year (Gahl, Thoene, & Schneider, 2002). Individuals are typically diagnosed in infancy and early childhood and face health challenges that include Fanconi's syndrome, growth delay, kidney failure, impaired vision, and numerous other systemic problems (Emma et al., 2014; Gahl, 2009). Similar to other rare pediatric-onset diseases, children with cystinosis and their families often have limited access to knowledgeable specialists or to accurate information about their disease and its treatments (Budysh, Helms, & Schultz, 2012). Over time, geographically and demographically diverse families affected by cystinosis have built a vibrant community for advocacy and support.

Before the availability of cystine-depleting treatment, children with cystinosis reached end-stage renal disease, requiring kidney transplant, around 10 years of age and faced early mortality by adolescence (Gahl et al., 2002). FDA approval of Cystagon in 1994 (Schneider, 1995) allowed for the slowing of disease progression (Kleta et al., 2004) and for survival into adulthood, with lowered rates of complications and delayed mortality reported in the United States and Europe (Gahl, 2009; Nesterova & Gahl, 2008; Schneider, 1995). Currently, the median age of survival is 22 to 29 years of age, depending on the age at which treatment is initiated (Brodin-Sartorius et al., 2012). However, there are now cystinosis patients in their 40s and 50s who are living with a spectrum of complications and reported quality of life. A rigorous treatment regimen includes a strict dosing schedule of oral cysteamine, which can result in challenging side effects such as gastrointestinal distress and a distasteful odor on the skin and breath. In addition, the daily regimen includes instillation of cysteamine eye drops every waking hour and taking large doses of mineral supplements and/or transplant immunosuppressants. Adherence to this regimen is correlated with improved outcomes later in adolescence and early adulthood (Gahl, Balog, & Kleta, 2007; Nesterova, Williams, Bernardini, & Gahl, 2015). However, the rigid timing of dosing and medication side effects often challenge adequate adherence during the adolescent years (Brodin-Sartorius et al., 2012). A recently approved extended-release medication (Pollack, 2013) appears not only to improve adherence and quality of life but also to preserve kidney function (Langman et al., 2014). These medical advances have transformed cystinosis from a fatal to a chronic disease (Doyle & Werner-Lin, 2015; Nesterova & Gahl, 2013).

### Living with a Child's Rare and Chronic Illness

Cystinosis is characterized by diagnosis in early childhood, a demanding treatment regimen, a chronic and slowly progressive course, and early mortality in the absence of treatment. Its recessive genetic form means siblings may be affected, may be carriers, or may be genetically unaffected. Given the disorder's psychosocial features (Rolland & Werner-Lin, 2006), evolution, and prognosis (Nesterova & Gahl,

2013), cystinosis shapes all aspects of family life, affecting the family's psychological, behavioral, educational, reproductive, and financial well-being (Cystinosis Research Network, Doyle, & Hammond, 2011; Spilkin & Ballantyne, 2007; Trauner, Spilkin, & Ballantyne, 2010). Despite advocacy group efforts and greater access to online resources (Beall, 2001; Ladd, 2015; Patsos, 2001), families of children with cystinosis might feel isolated from others who share their experience (Doyle, 2015). Patients now entering adulthood were diagnosed before or early on in the availability of cysteamine treatment. Their parents generally received dire prognoses, including the expectation that their child would die before reaching adulthood (Doyle & Werner-Lin, 2015).

Providers often consider health-related quality of life of children with chronic illnesses such as cystinosis by the barriers and deficits they report (Delgado, Schatz, Nichols, Appelbaum, & Trauner, 2005; Rajmil, Perestelo-Pérez, & Herdman, 2010). Yet, families adapt. They demonstrate resilience in the face of immense challenges (Dellve, Samuelsson, Tallborn, Fasth, & Hallberg, 2006; Nagy & Ungerer, 1990) and devise strategies to integrate aspects of normative development and illness demands into family life (McDaniel, Doherty, & Hepworth, 2014). These strategies are grounded in evolving philosophies about the place of illness in family life, and support coping, mastery, and survival. The recognition of the philosophies, values, and strategies that evolve or are chosen by families can help the health care team to support these patients—and their families—throughout the lifespan.

The primary study questions for this investigation focused on the transition to adulthood and adult-oriented care for individuals with cystinosis (see Table 1). Findings regarding the transition experience itself, and the importance of peer support, have been described previously (Doyle, 2015; Doyle & Werner-Lin, 2015). As a core category, which organizes the other codes and categories regarding participants' main concern (Glaser, 2002), participants "recognized the reprieve" that changing medicine has offered in extending the lifespan of cystinosis patients into adulthood (Doyle & Werner-Lin, 2015). However, the strategies put toward supporting the affected person's survival (through diagnosis, childhood, and adolescence) that emerged through the iterative process of coding went beyond the original sensitizing concepts of the study (emerging adulthood [Arnett, 2004] and health care transitioning). This iterative process and the Glaserian grounded theory tenets of avoiding preconception (Glaser, 2012) and trusting in emergence (Artinian, Giske, & Cone, 2009) allowed for the discovery of new "middle range" theory (a set of interconnected concepts that describe processes with greater specificity than working hypotheses but do not meet criteria for inclusive, grand level theory; Glaser & Strauss, 1967). Based on the nature of the data, the focus of this analysis is the individual perspective of parents and patients—adult patients reflecting on what their families had done to support their survival, and parents reflecting on behaviors and choices made to support their child's survival—which

**Table 1***Overview of Interview Guides for Adult and Parent Interviews and Focus Groups*

Interview Guide	Adults with cystinosis	Parents of Adults with Cystinosis
Transmutation of disease/illness (Feudtner, 2003)	How has having cystinosis changed as you've gotten older?  What changes in the treatment of cystinosis have impacted you?	How has having cystinosis changed as your child has gotten older?  What changes in the treatment of cystinosis have impacted them?
Emerging Adulthood (Arnett, 2000)	Do you think of yourself as an adult?  How do you define adulthood?	Do you think of your child with cystinosis as an adult?  How do you define adulthood?
Transitioning (AAP, AAFP, et al., 2011)	What are/were your concerns moving from pediatric to adult care?	What are/were your concerns moving from pediatric to adult care?

revealed strategies that supported cystinosis patients in reaching emerging adulthood, health care transition, and beyond. These strategies do not negate the difficulties and despair that rare and chronic illness can impose on patients and families, but suggest ways for patients, families, and clinicians to respond successfully to the demands of chronic conditions.

## Method

### Sampling and Recruitment

New York University's Committee on Activities Involving Human Subjects approved the study protocol, which originated in the Silver School of Social Work. Transitioning to adult-oriented care generally occurs between the ages of 18 and 21 years. Although preparation for health care transitions might occur before this, almost no literature has addressed the needs and experiences of adults with cystinosis. As a result, inclusion criteria required participants to be (a) 18 years or older with a diagnosis of cystinosis, or (b) a parent of an individual 18 years or older with a diagnosis of cystinosis. The majority of participants were initially recruited and screened online through 1 of 3 U.S.-based cystinosis advocacy organizations and their social media sites, whereas a few participants were recruited in person at three cystinosis conferences. Interested parties completed a brief questionnaire (available online and in paper format) to provide contact information

confidentially and to screen for inclusion/exclusion criteria. All participants received information about the study and completed informed consent in person prior to participation. Additional recruitment efforts continued online as well as through purposive and snowball sampling (Padgett, 2008).

### Data Collection

Data were collected via individual interviews and focus groups. Interview guides for focus groups and interviews were developed specifically for this study (see Table 1). The focus group and interview guides were developed with the help of key informants, including adult and adolescent members of the cystinosis community, parents, health care providers, and other researchers. Each participating adult with cystinosis completed a demographic questionnaire (age, gender, education, employment and income) and health history (age at diagnosis, transplant status, and age at transplant).

A researcher traveled to individual participant homes to conduct interviews with adult patients and their parents. The researcher also attended three national consumer-oriented cystinosis conferences to conduct focus groups with adult patients and with parents. Individual preference and availability dictated whether participants took part in an individual interview, one or more focus groups, or both. Parents participated with assent of their adult children (see Table 2). Data for adults with cystinosis and for parents were collected separately based on the study's aim of understanding the experience of transition to adulthood and adult-oriented care for each stakeholder.

The first author, a licensed clinical social worker, conducted six hour-long focus groups (see Table 3) at three cystinosis conferences that brought together patients of all ages, family members, health care providers and researchers. Separate focus groups were held with adult male cystinosis patients, adult female cystinosis patients, and parents; one focus group with patients of both genders was held at the third conference, with two repeating participants (see Table 3). Focus groups allowed for the collection of rich data through the group dynamic as participants influenced each other by their presence and by their reactions to what was said (Mack, Woodsong, MacQueen, Guest, & Namey, 2005). Focus groups are particularly well suited to communities, such as this rare-disease community, in which there is a preexisting "web of social networks" (Padgett, 2008).

Also during this time, the first author completed 17 individual semi-structured interviews with adults with cystinosis in addition to separate interviews with parents; some participants took part in both an interview and one or more focus groups. A crucial element of the study's design was the use of these in-person, at-home interviews that enabled study participation for a number of participants who could not travel to conferences for medical or financial reasons; this design ele-

**Table 2**

*Study Participants by Gender, Patient or Parent Role, and Data Collection Type (Focus Group, Individual Interview, or Both)*

Participants by gender, and data collection type	Focus group only	Individual interview only	Both individual interview & focus group	Total
Males with cystinosis	9	—	2	11
Females with cystinosis	3	5	2	10
Mothers	8	2	3	13
Fathers	8	2	—	10
Spouses	—	2	—	2
Total Participants	28	12	6	46 (from 22 families)

ment was a response to the Spilkin and Ballantyne (2007) study that noted the limitations of collecting such psychosocial data from cystinosis families via survey. Interviews generally took place in participants' homes and lasted 90–120 minutes.

Focus groups and interviews were audio recorded and subsequently professionally transcribed. In the tradition of grounded theory, field notes and memos were written immediately after each focus group or interview. Data collection continued until theoretical saturation was reached; that is, the point at which qualitative data collection and analysis yields additional phrases and incidents from participants but generates no new codes or categories (Glaser, 1978).

**Table 3**

*Focus Group Timing and Participants*

Focus group	Conference	Participants
Focus group 1	Conference 1	4 females with cystinosis
Focus group 2	Conference 1	7 males with cystinosis
Focus group 3	Conference 1	6 mothers of adults with cystinosis
Focus group 4	Conference 2	3 males with cystinosis
Focus group 5	Conference 2	5 mothers (1 repeat), 5 fathers of adults with cystinosis
Focus group 6	Conference 3	Mixed—3 males (1 repeat) with cystinosis & 2 females (1 repeat) with cystinosis

### Data Analysis

Substantive coding of transcripts was carried out in the grounded theory (GT) tradition of constant comparative analysis, as data collection and analysis was simultaneous and iterative (Charmaz, 2006; Glaser, 1978; Padgett, 2008). This methodology was chosen based on its frequent use in exploratory, qualitative health research, and particularly given its focus on conceptualization over description.

Data analysis began with line-by-line coding of the first three focus group transcripts to identify a preliminary list of codes. This coding included open and in vivo codes as well as focused codes addressing features of emerging adulthood (Arnett, 2004) and health care transitioning (American Academy of Pediatrics et al., 2011). Focused coding was also used to identify specific psychosocial and medical details, including education, work, relationship status, childbearing status, medical progression, and medical regimen. Researchers created a codebook listing code definitions and parameters, brief memos, and examples of each code, and then completed another round of coding to collapse and clarify emerging codes. Codes were grouped as (a) individual, family, and community contexts; (b) medical issues; (c) health care transitioning; and (d) emerging adulthood. Researchers met regularly to discuss emerging codes and categories, to revise interview guides, and update the codebook. The researchers used ATLAS.ti 7 (Scientific Software, 2013) to organize and store transcripts, codes, and memos.

The findings reported here, on parent and adult-child perspectives on family strategies for living with illness, emerged within individual, family, and community contexts. Findings specific to health care transitioning and emerging adulthood have been reported elsewhere (Doyle & Werner-Lin, 2015; Doyle, 2015). Phrases in *italics* denote codes that emerged through constant comparative analysis. Quotes are included to further illustrate codes and share the experiences of cystinosis patients and families with readers. When necessary in reporting findings, participants are described by age, gender, or parental role, but demographic information is used as rarely as possible to minimize the risk of inferred identification in this small rare-disease community (See Limitations).

### Trustworthiness

Criteria for assessing qualitative and grounded theory research include trustworthiness (Lincoln & Guba, 1986), credibility, usefulness (Charmaz, 2006), transferability, and confirmability (Padgett, 2008). Social work and health researchers provided peer debriefing (Padgett, 2008, 2009). The researcher participated in the Cystinosis Research Network's Adult Care Excellence initiative, the development and analysis of the Living with Cystinosis survey (2011), and the creation of the Bridges to the Future Transition Guide (2011); these opportunities provided prolonged engagement (Padgett, 2008, 2009) and insight into the concerns of the community. Comments from patients and families in advocacy organization newslet-

ters and social media have described themes similar to the concepts identified in this study, thus providing triangulation (Padgett, 2008, 2009) with publicly available information.

### Findings

The current study had 46 participants: 22 adults with cystinosis (12 men, 10 women) who were 18 years and older, and 24 parents (13 mothers, 11 fathers) of individuals with cystinosis (see Table 2). Most of the adult participants with cystinosis had one parent who participated in an individual interview and/or focus group, and several had both parents who participated. One pair of parents participated within a year of their adult child dying of cystinosis-related complications. Through constant comparative analysis, a set of behaviors or *strategies* (a theoretical code as identified by Glaser [1978]) emerged demonstrating how participants (both patients and families) negotiate their lives with cystinosis, acquire skills for managing the illness, and find a fit between normative developmental tasks and illness management—all of which enable them to *recognize the reprieve* that improved treatment has offered. These strategies emerged in the codes generated across all three contexts (i.e., individual, family, and community), and they entailed *educating, regimenting, pushing and protecting, connecting, coming to terms, and finding a fit*.

#### Educating and Regimenting

Parents had immersed themselves in the project of understanding disease mechanisms, treatments, and complications, and sharing this information with others in the life of the affected child (e.g., extended family, teachers), both at the time of diagnosis and throughout the child's changing medical trajectory (e.g., chronic kidney disease, dialysis, transplant, and post-transplant). Participant parents reported seeking out all the information they could find about the disease upon their child's diagnosis, *educating* themselves, other family members, caretakers and teachers, and eventually their child. When patients reached adolescence and young adulthood, they also began to take ownership of this information to prepare for greater responsibility in disease management. Participant parents described learning about cystinosis before the existence of the Internet, using limited search tools to find information that was "*dated and scary*." The explosion of Web-based information has enabled patients and families to access detailed and accurate information much more quickly, changing the information-gathering process during the lifetime of study participants.

Parents found ways (and the words) to teach their affected child about the disease. For some parents, explanations were given in small pieces, evolving to tell a child about changing medications or a hospital admission. As children grew older,

information about cystinosis became part of ongoing family conversations. As families became more experienced with illness management, parents and child (and then adolescent) *came to understand* crucial facts of the disease—what happens at the cellular level, how cysteamine works, how kidney function changes, and what medications need to be taken when. The following participant quotes were typical of the comments regarding educating children about cystinosis:

*Always very simply, based on his understanding. I mean we told him that he had cystinosis, that it was a disease that made his kidney sick, tried to do it a simple way . . . It's always something we talked about from the age he could talk.*

*This could happen; your kidneys could fail. [My parents] were always real with me with what cystinosis could do. I could die. They were very supportive as well and educative about it.*

*We just tried to explain to her as much as possible what cystinosis is. I think we're all on different curves as far as our learning and being able to describe what it does and how it works and it's a genetic thing . . . I think she knows all that stuff. She learns a little bit at a time, and it's always going in the bank. I may be discounting her. She probably can verbalize it much better than I realize she can.*

Just as children and adolescents with cystinosis *become acclimated* to their medical regimen, they also became acclimated to having the disease and having to explain it to others. Some participants described having knowledge of cystinosis for as long as they could remember:

*I always knew. It's weird. I know we went to a conference in Boston, and I was 7. And I knew what it was. I have a disease, and it creates crystals in my body. And everyone would ask, "Why do you take so much medicine?" Because I would go to the nurse in school. And I would explain to people. I grew up knowing what it was.*

*I guess I've always learned about cystinosis—I've been very fortunate to come to conferences and learn from the experts themselves. At conferences, [my parents] always encouraged me to attend the sessions even when I was really young.*

In *educating themselves and coming to understand* the disease, members of the cystinosis community—including parents, affected individuals, or other caregivers and family members—used both in-person and online social networks and resources. Patients and families acquired information via advocacy organizations and through PubMed, from attending conferences (which typically include presentations of complex research findings), and from the processes of *comparing notes* and *mentoring* that goes on both online and in person (Doyle, 2015). The level of health literacy in

this rare-disease community was particularly striking, as were the partnerships of providers, researchers, advocates, and industry that made information available and understandable. Individuals and families gained a modicum of control over the illness as they educated themselves, shared information with one another, and communicated effectively with health care providers and researchers.

Another step in gaining some sense of control over the illness was the process of *regimenting* that parents embarked upon, a code that describes creating systems and structures to organize the care of a child with cystinosis with the aim of stabilizing care within daily life (Doyle & Werner-Lin, 2015) and to somehow normalize the task burden of the children's care. One adult participant described how regimenting was reinforced: "*Taking medicines is the same normalcy as brushing your teeth in the morning. It's just something you do, but it's not something that takes you away from doing Scouts, or going to school, or church, or whatever else.*"

When children with cystinosis are young, parents create a routine of administering medications, supplements, and nutrition necessary to sustain their child's health. This strategy was not only revealed during interviews but also was visible at conferences (where focus groups and interviews were held) where gastrostomy tube supplies and medication carriers littered the tables around mealtimes. Parents enforced the idea that both the taking and timing of medications was not flexible, as pharmacokinetics and risk of transplant rejection make dose timing critical (Grimm, 2012). *Regimenting* was subject to creativity, but not flexibility. As illustrated by the following comments, adults with cystinosis acknowledged the creativity required of parents to get medications and mineral supplements into their small children:

*I think I started swallowing pills when I was 4, because I hated the grape juice mixed with pills. My dad would dress up as Vita-Man, with a mask and stuff, and I'd hide behind the couch. Dress up and make Dad look really stupid.*

*I remember my parents trying to mix the [liquid medication] with stuff. Whatever they would mix it with, I still hate — like pineapple juice, I can't drink pineapple juice because they would try to mask it, and there was no masking.*

As parents educated themselves and came to understand the illness and its treatments, they found ways to convey gentle but straightforward information to their children about why the medications were vital to their survival, as one parent ardently described:

*One thing that we always, always, always, back to the beginning, when we were trying to get him to understand the importance of medications, we always reiterated what would happen, that there are consequences if you don't do this, and this might be what it does.*

### Pushing and Protecting

When possible, parents encouraged independence and risk-taking among their children with cystinosis. These parents expected children with cystinosis to accomplish some, all, or perhaps even more, than healthy siblings or same-age peers, particularly during secondary education. While *pushing* children to undertake typical and illness-related tasks, parents also valued *protecting* children from the exigencies imposed by cystinosis and its treatments. The delicate balance between *pushing* and *protecting* aimed to promote the best possible health in the least-restrictive environment. To promote normalcy, some parents refused to let their children with cystinosis occupy a *sick role* (Charmaz, 1999; Parsons, 1951). Parent participants described trying hard to maintain the same discipline and responsibilities at home for siblings with and without cystinosis:

*The hard thing was when other parents would tell us that she couldn't do things, or often it was at school. But it's not held her back, because I didn't let it. I didn't let it be an excuse to being a productive human being in this world. It's not an excuse. You happen to have it, take your medicine, do it well.*

*You're not going to tell me you're sick and can't do your homework. Oh, no, you're a bright kid. There was just no compromise on any of that. We wanted her to be independent. We wanted her to succeed.*

*She had this little clique of girls that were all in dance, and one of the moms called me, and she said, "Do you know what your daughter's telling everybody at dance today? That she's getting a kidney transplant next week," and I said, "She is." "Well, she's not sick?" and I said, "Well, she is sick, but she doesn't want to be different."*

Parents invested heavily in keeping their child healthy enough to reach adulthood. To support future-oriented planning, parents felt a responsibility to give their children tools for survival in the real world, with expectations of, for example, finishing school or getting a job. The use of the word *tools* refers not only to coping strategies but also to practical skills like medication regimenting and technological fluency. As the following quotes demonstrate, through the combination of *pushing* and *protecting* their child with cystinosis, parents set expectations as well as set the stage for *defying expectations*:

*I worry less about her surviving in this world because I know she's got the skills to do it.*

*She doesn't venture very far because she has a terrible sense of direction. Now we have GPS. She can punch in an address, and she's gone, and she's not afraid. You let them go, and you give them the tools to survive in this world, whatever that is, like a GPS.*

*[My parents said] I don't care what you do, as long as you go to college and do stuff.*

Even with parents *pushing*, and children with cystinosis *defying expectations*, parents and young adults alike recognized the need for extra health care services, parental attention, accommodations, and/or special education services at school. While *pushing* children to try harder or do more, parents were simultaneously *protecting* them, as they identified and set limits, aware of losses caused by the disease. For example, visual processing difficulties created concerns around driving for one family: “*She has problems processing the traffic. If she gets in multiple lanes of traffic or at crossroads, she can't process which car is doing what when, and she just—brain overload—freezes, so we work with her.*”

A father spoke about the difficulty of telling his son he could no longer compete in a favorite sport:

*He started wrestling when he was 6 or 7 and wrestled until he couldn't wrestle any more because of his kidney failure, his sophomore year in high school. We found a [doctor] who said that his nephrologist was being too protective and let him wrestle. Neither one of us could take that chance. It's too big a risk, and sometimes the answer is no, and this was no. It was a big hurdle, and once he got past it, he did really well. The school handled it real well for him.*

Later in their lives, an element of *coming to terms* and *finding a fit* for individuals with cystinosis was finding a comfort level in pushing and protecting themselves. Young adults pursued (and adapted) goals and dreams while advocating for the supports and services they needed.

### Connecting

*Connecting* is an umbrella term for the many ways families and individuals with cystinosis connected with sources of social support who shared their experience, be it with others affected by cystinosis; other rare, metabolic, or renal disorders; or dialysis and transplant. Many but not all study participants were active members of the cystinosis community through participation in patient advocacy organizations, annual family meetings, or online social media and support forums. Behaviors demonstrating the ways in which participants connected and live with illness in the context of the cystinosis community have been described previously (Doyle, 2015).

For families with chronically ill children, extended family members can be valuable sources of support. For example, at conferences, the first author met extended family (e.g., aunts, grandparents, cousins) who had clearly become part of the cystinosis community, helped care for the affected child, took care of siblings, or engaged in fundraising. However, at times, extended family members proved

unable or unwilling to understand the disease or provide material help to affected families (Patterson, Garwick, Bennett, & Blum, 1997). Sometimes extended family members would instinctively pull away to avoid their own grief. One mother described calling members of their “cystinosis family” before calling extended (biologic) family when her daughter had a health challenge or when she, as a parent, needed support. She described the special connection with other families affected by cystinosis: “*In many respects, I feel closer to this group of people than I do biological family. They [cystinosis family] know us in ways that our own family probably can't know us.*”

Another participant described the opportunity to feel *comfortability*, that is, to relax, and to feel at ease being in contact with others living with cystinosis:

*Just having those connection lines and the comfortability of being able to just pick up the phone and call somebody whether any issue, and even if it's not an issue, you being able just to have a conversation and relate exactly what you've gone through in the different generations is really helpful.*

Although many participants reiterated that their illness does not define them, participating in the cystinosis community provided a safe environment to be with people who “*just get it,*” to talk freely using a cystinosis-specific vocabulary, and to be less guarded.

### Coming to Terms and Finding a Fit

In raising a child with cystinosis, families established a philosophy about how life was organized around managing illness, rather than sacrificed to illness. If *educating* and *coming to understand* describe the process of learning about the disease and its management, *coming to terms* was the negotiation of a balance between illness and other aspects of life, such as family roles, chores, activities, and expectations for the future. As individuals with cystinosis became adults, they incorporated the terms set by their family and renegotiated those terms for themselves.

Participant parents described a range of approaches to negotiating the demands of the illness with those of daily life and development. *Finding a fit* remained a fluid and ongoing deliberation about integrating illness into family life or centering family life on illness symptoms and treatment. Parents instilled values they perceived would help their children maintain a balanced perspective on their illness, which included acceptance, independence, resolve, patience, resilience, and flexibility.

*One of the lessons we've tried to teach is we've got to do this on our terms to some degree, because you've got to have a life of some kind. Sometimes the medical provider's going to have to wait a day or two, just because — it's your life.*

*It's made our life go in a different direction than it may have been but, honestly, I think maybe it's probably been a better direction than it would have been otherwise. You just learn so many other things that you might have taken for granted or not realized. You learn to value things, what's really important and what isn't.*

*That's the way we, as a family, have faced each one of these things, is deal with the problems that are closest at hand. Don't get crazy about the ones that aren't here yet. Do what you can to prepare for it, but, again, don't let it ruin or control your life.*

Participant parents described a range of approaches to *finding a fit*—whether illness fits into family life or whether family life is organized around one member's illness.

*Cystinosis doesn't control my life or her life. It fits into our life.*

*I had three kids. He was the third. I wasn't going to sacrifice all of them to cystinosis, so we were not going to make our life focus [on] cystinosis.*

*She was diagnosed at 14 months. My stubbornness kicked in, because I said, "we've had this child for over a year. She's fun. She's beautiful. We're not going to live cystinosis as a life. We have a life, and we're going to live it.*

*We haven't learned to fit cystinosis in our life. We've learned to fit life around the cystinosis. Life will happen when she's having a good day, when she's feeling up to it.*

*Finding a fit* also meant families discovered creative ways to include fun, play, and happiness in daily life.

*We realized real early—to have any type of life—we had to maximize the moment. No matter where we were going, what we were doing, we would always say, "Hey, we're back together. Let's go." And it would be an adventure.*

Adult participants with cystinosis described carrying forward the values and attitudes towards illness instilled by their families. Some observed that, in light of their illness, other things in life were not so serious, and other situations were much worse.

*Once you get to a certain age you're tired of dealing with that smell and everything, but once you get more mature, you realize, life has problems, but you've got to deal with it. There're a lot of people that's a lot worse off.*

*Some itty-bitty thing, like, oh, I got a bumper thing with my mom's car, and I'm like, no big deal. I mean there are things that are a lot worse in the world.*

*Coming to terms* changed over time as individuals grew up with cystinosis. They faced personal, social, and medical challenges that arose with the disease and renegotiated those terms for themselves. Many young adults in the study echoed thoughts of *valuing and living in the now*, as they recognized their mortality, clarified their values, and made deliberate choices about how they wanted to live. Adult participants spoke about how they had *come to terms* with the disease in their lives by developing and adapting the philosophy established by their families:

*It's not all about having the disease. You're more than just that.*

*Ten years from now, I want to look back, and I want to know that I moved my life forward and I lived my life to the fullest. But I also want to know that I did everything I could.*

### Discussion

When using a strengths-based approach with such families, and recognizing the evolution of such strategies over time, the challenges to the family cannot be understated. A 2007 study of parents of children with cystinosis found that 42% of respondent fathers and 75% of respondent mothers endorsed that the disease affected their family negatively, particularly in terms of time, worry, and financial strain; one mother in that study described how the disease “consumes our lives” (Spilkin & Ballantyne, 2007). Findings from Spilkin and Ballantyne’s 2007 study and from other studies of family processes in pediatric rare-disease groups serve as an important reminder to look beyond the perspective of the primary caregiver when seeking to understand the impact of a child’s illness on the family. Although most participants with cystinosis advocated great cohesion with (and gratitude toward) their parents and siblings retrospectively, they expressed clear differences of opinion about parenting, treatment adherence, and future plans. The process of educating themselves and important others about cystinosis provided a vital pathway for families, and later the patients themselves, to reassert control over their lives. As previously noted, a study of pediatric patients with cystinosis and their parents (Wolff, Theilen, & Ehrich, 1992) found greater adherence among patients who felt actively informed and able to influence at least some aspects of their treatment, and who felt that their efforts to cope were recognized by their providers.

The strategy of *regimenting* aligns with findings from a previous study of quality of life with cystinosis (Trauner et al., 2010). Trauner and colleagues suggested cystinosis families rely on structure and organization by setting rules and procedures to address the demands of the illness. Adherence with medications has been clearly linked to improved outcomes for cystinosis patients (Brodin-Sartorius et al., 2012) and kidney transplant recipients. As children mature, regimenting extends

to greater self-management, but parental surveillance improves adherence during adolescence and on into adulthood (Ellis et al., 2007). Emerging adults and adults with cystinosis find new ways of regimenting that fit their lives as they move outward from their families of origin to higher education and work settings, and to new families of choice.

As a test of transferability, the first author has presented these strategies at meetings for parents of children with cystinosis and for adults living with cystinosis, as well as those affected by other rare conditions such as atypical hemolytic uremic syndrome and focal segmental glomerulosclerosis. Feedback from attendees suggests that these strategies ring true across a diverse group of families who are able to attend national or regional consumer conferences (often made possible by travel and patient education grants).

Ongoing engagement with the cystinosis community presents further anecdotal evidence that families who use the described strategies to a greater extent feel more control over the impact of disease in their lives and less guilt over aspects of disease progression. Since this study's completion, two participants (and a number of other adult patients in the cystinosis community) have died of disease-related complications, including advancing myopathy. Families have demonstrated resilience in the face of these losses due in part to the strategies used to extend their loved one's lifespan and support them throughout their trajectory with cystinosis. For example, such strategies include staying connected with the disease community and continuing to help raise awareness, raise funds, or advocate for research.

### Limitations

The specific population, sample size, qualitative nature, and methods of this study provided rich data regarding the experiences of adults with cystinosis. Recruitment efforts might not have captured the full range of family strategies used to address the challenges of cystinosis, particularly for those who are disconnected from the disease community by choice or circumstances. Although many self-selected participants were in "good" health, most faced disease complications—primarily muscle-wasting—that can, over time, affect movement, speech, swallowing, and respiration. Whereas some families were confident about the future, others had great concern for their adult child's future health and autonomy, even while they celebrated their successes so far.

The findings presented here might or might not be comparable to the experience of individuals with other chronic and rare diseases or, given changes in available treatment, to other birth-cohorts affected by cystinosis. Additionally, the experience of living with cystinosis, and of transitioning to adulthood and adult-oriented care, might be quite different outside of the United States or other developed nations, with limited access to cystine-depleting therapy and differences in organ

allocation for transplant (Tsygin, Kagan, Kartamysheva, & Levchenko, 2011; Vaisbich & Koch, 2010). This study's primarily Caucasian sample might not thoroughly reflect the ethnic and racial demographics of individuals with cystinosis in the United States or worldwide, although the mutation has been identified more frequently in those of Northern European descent. However, the study sample was diverse in terms of socioeconomic status, education, and employment.

The small size and high level of interconnection throughout the cystinosis community increases challenges to anonymity and confidentiality, including the risk of inferred identification (Anastas, 2004). The thick description made possible through this study presents a particular challenge between dissemination of findings with meaningful detail and privacy of individual participants who are well known to each other, to providers, and to other researchers. Classic grounded theory methodology helped to organize data around codes and concepts rather than around descriptions of individual participants or families.

### Implications

The identified strategies—*educating, regimenting, pushing and protecting, connecting, coming to terms, and finding a fit*—highlight prime areas for psychosocial intervention and support by health care team members engaged with patients and across the family life cycle, whether newly diagnosed or in the “long haul” with this chronic condition (Rolland & Werner-Lin, 2006). As understanding of rare and genetic childhood diseases expands and new treatment options become available, health care providers and multidisciplinary teams should consider regularly assessing the strategies used by families to manage illness. These strategies must adapt as treatment protocols evolve and as children survive into adulthood to face new, and perhaps unexpected, developmental tasks. The strategies described by participants might help frame the expectations of families and organize how resources and support are offered. A dynamic framework for understanding these strategies must incorporate a developmental perspective to provide appropriate tailoring of interventions and communication to the individual and family living with cystinosis or similar rare and chronic conditions.

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