

# Behavior in Children With a Chronic Illness: A Descriptive Study of Child Characteristics, Family Adjustment, and School Issues in Children With Cystinosis

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*When caring for a child with a chronic illness, parents, physicians, and other medical personnel often focus their attention on treatment of the medical illness. The child's and family's coping and mental health are often secondary to medical management of the disorder. Cystinosis is a genetic metabolic disease that affects multiple organs, and specific aspects of cognition and behavior. The present study examined behavior and adjustment in a sample of families and children with a chronic illness, cystinosis. The Cystinosis Behavior Questionnaire was administered to 63 parents of children and adolescents with cystinosis (ages 2–17 years). The questionnaire was comprised of*

*both open- and closed-ended questions that probed areas of child characteristics, family adjustment, school performance, and intervention for mental health issues. Parents reported many areas of strength and difficulty within their child and family. Interestingly, however, very few of the families had sought out intervention for behavioral and/or adjustment issues. In better defining behavioral and adjustment issues in cystinosis families, the current findings may prompt greater awareness in individuals caring for and/or working with a child with cystinosis.*

**Keywords:** cystinosis, chronic illness, coping, behavior, adjustment

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Cystinosis is a rare genetic autosomal recessive disease with an incidence of between 1 in 100,000 and 1 in 200,000 (Adamson, Anderson, & Gahl, 1989; Gahl, Thoene, & Schneider, 2002). It is a metabolic disease of lysosomal cystine storage, in which the amino acid cystine accumulates within the lysosomes of all cells in the body. The earliest and most clinically affected organ is the kidney. As individuals with the disorder age, other organs, such as the thyroid, pancreas, cornea, and brain, become affected as well. Owing to advances in the medical

treatment of the disorder (cystine-depleting medication, kidney dialysis, renal transplantation), individuals with the disease are now living into the third and fourth decades of life. The purpose of this article is to shed light on the numerous nonmedical challenges faced by children with cystinosis and their families.

Theoretically and practically, for families with a child who has a chronic illness, the demands of the illness (e.g., the course, level of incapacitation, outcome, etc.) interact with the dynamics of the family system (e.g., the family's beliefs, communication, adaptability, etc.) to result in the mastery, or lack thereof, of the issues presented to the family by the disorder (Rolland, 1994). The central role of the family in our understanding of the biological, psychological, and social (i.e., biopsychosocial) demands of chronic illnesses is highlighted by the family systems-illness model (Rolland, 1994). The biopsychosocial model proposes that for families to optimize their functioning in the face of a chronic disorder, they must have a basic understanding of the demands of the illness (practical and emotional) and how these demands may change over time. Moreover, the family unit, and the individuals in the family, must be able to adjust their "style" to continue to meet the changing demands of the disorder. Although the importance of understanding the psychosocial aspects of cystinosis cannot be underestimated, there is a paucity of such information.

A useful starting point in understanding the biopsychosocial demands of cystinosis is to characterize the disease landscape. Using the family systems-illness model (Rolland, 1994) to define the psychosocial typology of cystinosis, the disease is gradual in onset (e.g., most children are diagnosed after weeks/months of failure to thrive), progressive (i.e., health deteriorates over time), constant (i.e., there is no period in which the symptoms remit), non-incapacitating (i.e., in the early stages of the disease, individuals with cystinosis can

function in day-to-day life with treatment; as the disease progresses, many individuals become incapacitated), and is possibly fatal and/or results in a shortened life span (i.e., with current treatment the life span can be into the third or fourth decade of life for some individuals). These illness characteristics create their own unique set of practical and affective demands. For example, a child with cystinosis may be given the diagnosis after months of failure to thrive and life-threatening illness (e.g., a 2-year-old child reduced to his or her birth weight), the diagnosis and its implications often throw the family into crisis, the symptoms are constant with no remission, and the medication regimen is around the clock (i.e., medication must be given every 6 hrs. throughout day and night). Furthermore, the intensive, ongoing treatment regimen requires extreme collaboration on the part of the family and can lead to exhaustion and frustration through the course of the disease.

In addition to the illness typology, the time course of cystinosis must also be considered. According to Rolland (1994), regarding chronic illnesses of all types, there are three phases that families go through: (a) crisis, which includes symptoms, diagnosis, and the initial adjustment period; (b) the chronic "long haul"; and (c) the terminal phase, including death (Rolland, 1994). Because the demands of each phase differ, the family (and individuals in it) must adjust its perspective and behavior to adequately address the challenges that each phase presents. Other factors that contribute to the biopsychosocial characterization of the illness include the meaning of the illness (e.g., in terms of control or stigma) and historical data about the illness (e.g., treatment, mortality). In terms of cystinosis, control is minimal to nonexistent, as this is a metabolic disorder that has no cure. Individuals face stigma because they have noticeable physical characteristics including short stature and photophobia, Cushingoid features if they have under-

gone renal transplantation, and strong body and breath odor due to medication side effects. Because cystinosis is a rare "orphan" disease, and one from which children have historically died in infancy or within the first decade of life when left untreated, there is relatively little consistent information available regarding the long-term outcome for individuals with cystinosis (e.g., with the advent of new treatments, the current expected life span is unknown).

Cognitive status is another variable that affects both the individual with a chronic disease and the caretaking demands on the family. Individuals with cystinosis have a high incidence of central nervous system abnormalities, as well as specific neuropsychological findings that may affect psychosocial adjustment. The cognitive profile of individuals with cystinosis appears to be generally intact overall cognitive functioning with specific areas of cognitive decrement. Overall intelligence has been found to be in the average range (Trauner, Chase, Scheller, Katz, & Schneider, 1988). However, deficits have been observed in the areas of visuospatial skills (Ballantyne & Trauner, 2000; Nichols, Ballantyne, Hodge, & Trauner, 1990; Scarvie, Ballantyne, & Trauner, 1996), visual memory (Nichols et al., 1990; Schatz, 2002; Trauner, Chase, Ballantyne, Tallal, & Schneider, 1989), tactile recognition (Colah & Trauner, 1997), and academic/mathematical skills (Ballantyne, Scarvie, & Trauner, 1997). This particular cluster of cognitive weaknesses may result in learning differences or difficulties in everyday life (e.g., learning to drive, independent living).

To assess the sociobehavioral characteristics of children and adolescents with cystinosis, an article published by our laboratory (Delgado, Schatz, Nichols, Appelbaum, & Trauner, 2005) utilized a widely used and well-standardized quantitative parent report questionnaire (the Achenbach Child Behavior Checklist; Achenbach, 1991). Compared with healthy controls, individu-

als with cystinosis had a significantly higher incidence of behavior problems, which was reflected on the Total Problems and Internalizing Problems summary scales specifically. Examination of the individual clinical scales revealed differences from controls on the Social Problems, Somatic Complaints, Attention Problems, Thought Problems, and Aggression scales. Anecdotally, parents of children with cystinosis have expressed concerns regarding their child's behavior and adjustment.

In contrast to the quantitative Child Behavior Checklist study, the present study aimed to further characterize the "personality" of cystinosis by providing a qualitative description of behavior problems and family adjustment in cystinosis and by capturing, in narrative form, the issues that these families face. We used a Cystinosis Behavior Questionnaire (CBQ) developed in our laboratory to further probe areas of behavioral function, dysfunction, and concern for these children and families. This questionnaire was designed to examine child characteristics, family adjustment, and school issues. Rather than asking closed-ended questions, as does the Child Behavior Checklist, the behavioral questionnaire included both open- and closed-ended questions, and it gave parents the opportunity to share their thoughts and feelings about their chronically ill child, as well as the strengths and difficulties in their families.

## METHOD

### Participants

CBQ data were analyzed for 63 children and adolescents with cystinosis. The questionnaires were completed by the primary and secondary caregivers of these children. The primary caregiver was mother ( $n = 58$ ), father ( $n = 4$ ), or grandfather ( $n = 1$ ). The secondary caregiver was father ( $n = 35$ ), stepfather ( $n = 2$ ), mother ( $n = 1$ ), grandmother ( $n = 2$ ), grandfather ( $n = 1$ ), aunt ( $n = 1$ ), or sibling ( $n = 1$ ). Twenty

questionnaires were not completed by a secondary caregiver. The individuals with cystinosis ranged in age from 2 years to 17 years, with a mean age of 8.5 years, and were in the chronic stage of the disease (i.e., the crisis stage [diagnosis and initial adjustment period] happened before the completion of the questionnaire and no individuals were in the terminal stage of the disorder). Questionnaires were completed on 29 boys and 34 girls, which represents the majority (>90%) of families who received this questionnaire. Participants were identified through our studies of cognitive development in cystinosis, as well as through the National Cystinosis Foundation. Parents individually completed the questionnaires and mailed them to our laboratory.

Participants were part of a larger study of cognition in individuals with cystinosis. Informed consent was obtained for all individuals before participation in the study according to Institutional Review Board procedures.

### Measure

In the absence of an existing standardized questionnaire to assess qualitative aspects of behavior and adjustment in cystinosis, the CBQ was designed in our research laboratory and used in the present study. The questions on the CBQ were designed to tap areas of child and family functioning that may potentially be of concern in families affected by cystinosis and were based on our experiences and discussions with parents who had previously participated in our research studies. One part consisted of 10 open-ended questions about the child's illness, parental stress, and family adjustment that were completed independently by both the primary and secondary caregivers of the child. Another part consisted of 13 open-ended questions that probed the areas of tantrums and mood swings (a particular area of concern for parents of children with cystinosis) completed by the primary caregiver. A final

part, also completed by the primary caregiver, consisted of 52 statements that were answered on a 3-point Likert response scale, with 0 = *rarely or not true*, 1 = *sometimes or somewhat true*, and 2 = *often or very true*. Questions on this part encompassed the areas of child characteristics, family adjustment, school performance, and intervention for mental health issues. Two main scales, created on the basis of the items' face validity, were termed *Child Characteristics and Adjustment* (28 items) and *Family Characteristics and Adjustment* (17 items). The internal consistency of the two scales was good, according to convention (Haugland & Wold, 2001), with alpha equal to .83 for the Child scale and .88 for the Family scale. These are quite favorable alpha levels for the assessment of a multidimensional construct such as overall child and family characteristics and adjustment.

On this part of the CBQ, 16 items were worded such that higher scores represented a greater degree of a positive attribute (e.g., "takes an active part in family affairs"), whereas 31 items were worded such that higher scores represented a greater degree of a negative attribute or dysfunction in the child, family, or school setting (e.g., "has a strongly defiant personality"). In addition, 1 item probed use of interventions (e.g., "seeing a therapist"). Four additional items on the questionnaire were not used in the current analyses because of their irrelevance to the focus of this article. All data (both opened and closed ended) were coded and reliability checked by an independent rater to ensure validity and reliability.

It should be noted that characteristics in Figures 1 and 3 are all worded positively and characteristics in Figures 2 and 4 are all worded negatively. On the questionnaire, questions were randomly worded to the negative or to the positive, and all were put on the same scale (i.e., in the same direction) for analysis purposes. Figures 1–4 were derived in the following manner:

Once items were on the same scale, the ratings of "often or very true" and "sometimes or somewhat true" were combined to represent whether a child demonstrated the particular trait or characteristic. Items receiving the highest and lowest endorsements were then rank ordered into the positive and negative charts.

## RESULTS AND DISCUSSION

Overall, examination of parent responses on the CBQ revealed numerous strengths and weaknesses in the areas of child characteristics, family adjustment, and school issues. This study was meant to provide a more qualitative description of some of the behavioral issues quantitatively documented in children with cystinosis (Delgado et al., 2005), as well as to address anecdotal concerns of parents regarding family stress and school difficulties. The need for a greater understanding of behavior in children with cystinosis is exemplified by the concerns of one mother:

It [cystinosis] is a sadness that I carry with me. I try not to let it affect our lives. Since K is our oldest, I am constantly asking if her behavior is normal or if it is affected by her cystinosis." (female, age 6)

### Child Characteristics in Cystinosis

Figures 1 and 2, respectively, illustrate the positive and negative characteristics parents reported in their child. Parents of children with cystinosis reported many positive qualities in their children (with exact percentages of responses provided). Parents almost unanimously reported that their children often or sometimes are not depressed, have a good sense of humor, handle their health problems maturely, have a pleasing personality, and are sensitive to others. On the other hand, parents also reported that their children have several negative traits. Most notably, more than 50% of

parents reported that their children often or sometimes get their feelings hurt easily, do not enjoy life, are not self-reliant, are strongly defiant, and are very irritable.

Sixty-five percent of parents reported that their children with cystinosis have occasional outbursts of temper or severe mood swings. Notably, 36% of parents believe that their chronically ill child's behavior is more intense than that of other children. In terms of the characteristics of the temper tantrums or mood swings, 52% of parents reported that the tantrums happen at home; that temper tantrums usually happen when the child does not get his or her way or a misunderstanding leads to frustration; and that tantrums are more likely when the child is tired. The tantrums consist of crying, yelling, arguing, and irritability. Descriptions of the tantrums from parent responses include the following: "argumentative," "stubborn," "snotty," "mean," "intense outbursts," "crying and screaming," "says hurtful and bad words," and "flies off the handle." Parents reported that reasoning with the child usually makes it worse, whereas ignoring it, time outs, and eating or sleeping makes it better. Thirty-nine percent of parents reported that family members become mad, impatient, or tense as a result of these temper outbursts. Occasional outbursts of temper or severe mood swings were not limited to the young children in our sample; when responses only for children age 8 years and older were examined ( $n = 35$ ), temper tantrums and/or mood swings were endorsed by 54% of parents.

These findings on tantrums in cystinosis are extremely noteworthy and, to some extent, consistent with the literature on behavior issues in chronically ill children. Although children with cystinosis have some behavioral strengths, they also have numerous behavioral issues that would be taxing to any family. In looking at Figures 1 and 2, as well as the open-ended informa-

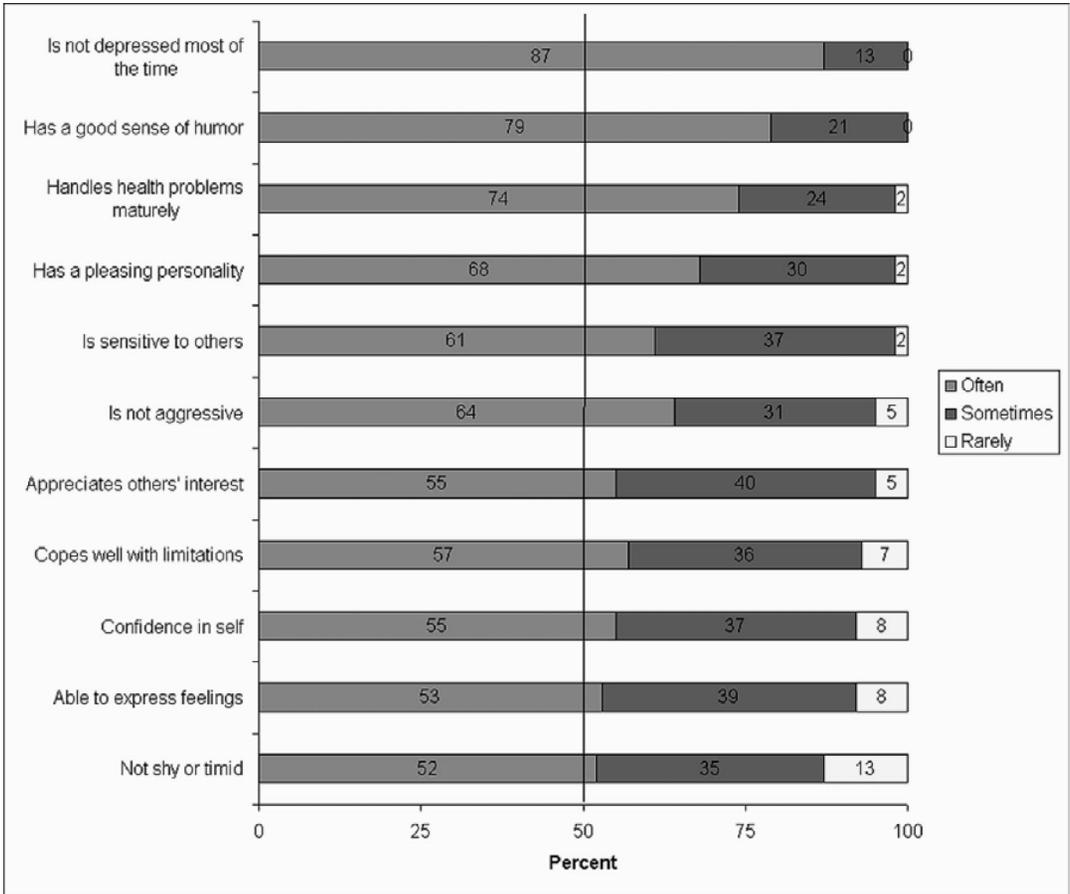
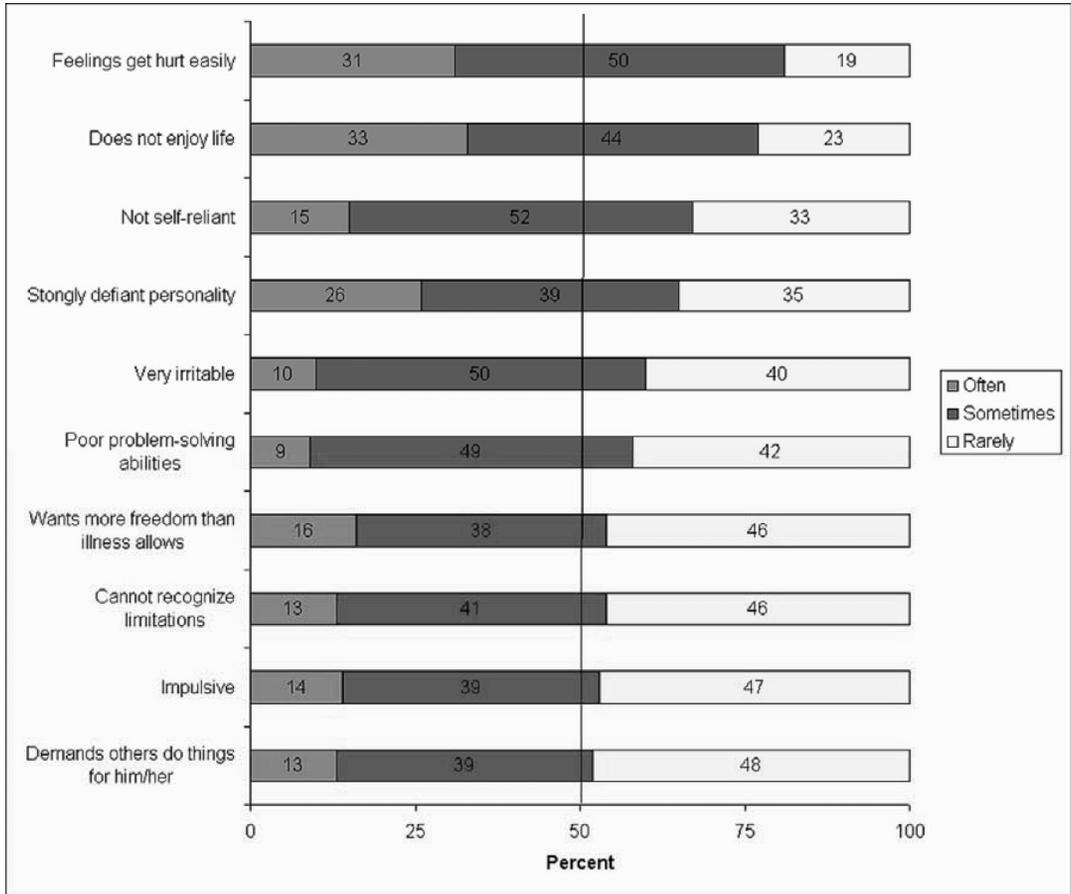


Figure 1. Percentage of Parents Responding “Often or Very True,” “Sometimes or Somewhat True,” or “Rarely or Not True” to Positive Child Characteristics on the Cystinosis Behavior Questionnaire.

tion on tantrums, one could speculate that the negative behavioral characteristics (e.g., getting feelings hurt easily, not enjoying life, very irritable, defiant) coupled with poor problem-solving skills and limited ability to express feelings could certainly lead to an increased incidence of tantrums. The behavioral problems endorsed by caregivers, along with intense tantrums, may bring about family adjustment issues. The cause of these behavioral difficulties in children with cystinosis is unclear; they may possibly stem from central nervous system abnormalities and/or a metabolic process. At this time, it is also

unclear whether the particular behavioral difficulties noted are disease specific or occur frequently in chronic illness in general. However, our prior research has indicated that children with cystinosis show increased behavioral problems when compared with both a typically developing group and a chronic disease control group (cystic fibrosis; Delgado et al., 2005). The impact of these behavioral issues goes beyond just the child’s psychological well-being and may also influence many other areas of the child’s life (Lavigne & Faier-Routman, 1992; Schmidt, Petersen, & Bullinger, 2003; Varni, La Greca, & Spirito, 2000).



*Figure 2.* Percentage of Parents Responding “Often or Very True,” “Sometimes or Somewhat True,” or “Rarely or Not True” to Negative Child Characteristics on the Cystinosis Behavior Questionnaire.

### Family Adjustment in Cystinosis

The families of children with cystinosis face numerous challenges, including special care demands (e.g., medication regimen), frequent doctor appointments, possible hospitalizations, and the psychological stress and worry prompted by having a child with a chronic medical illness. Even under these constant stressors, parents of children with cystinosis reported numerous strengths within their family. For instance, the vast majority of respondents reported that their children with cystinosis take an active part in family affairs, that they fit into the family social group, and that there is not a lot of anger in the family.

See Figure 3 for a depiction of family strengths.

However, as shown in Figure 4, parents clearly reported numerous family stresses, including worrying too much about their child’s health, taking on an inordinate amount of responsibility, doing things for their child that he or she should be able to do on his or her own, and having too much of family life revolve around the child with cystinosis. In addition to the special care demands of having a chronically ill child, these family adjustment issues can have an impact on the family unit.

In addition to the stressors within the family, some interesting differences of

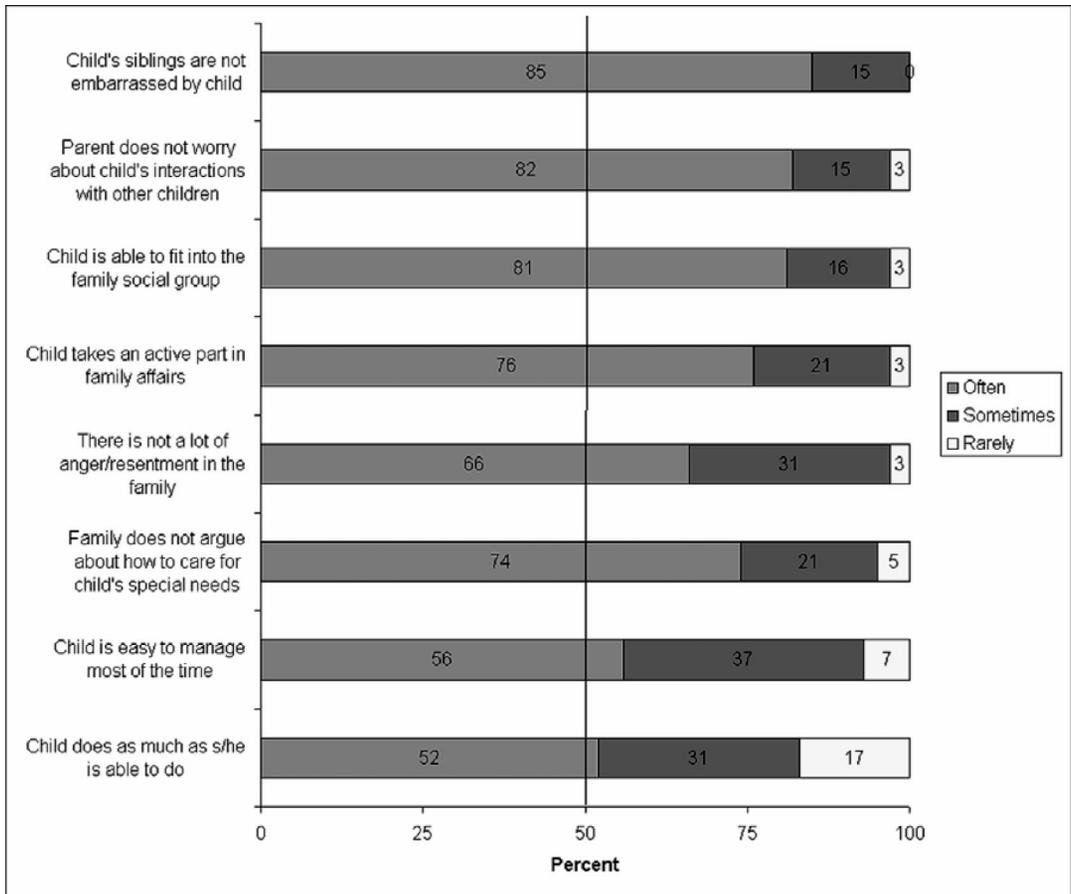
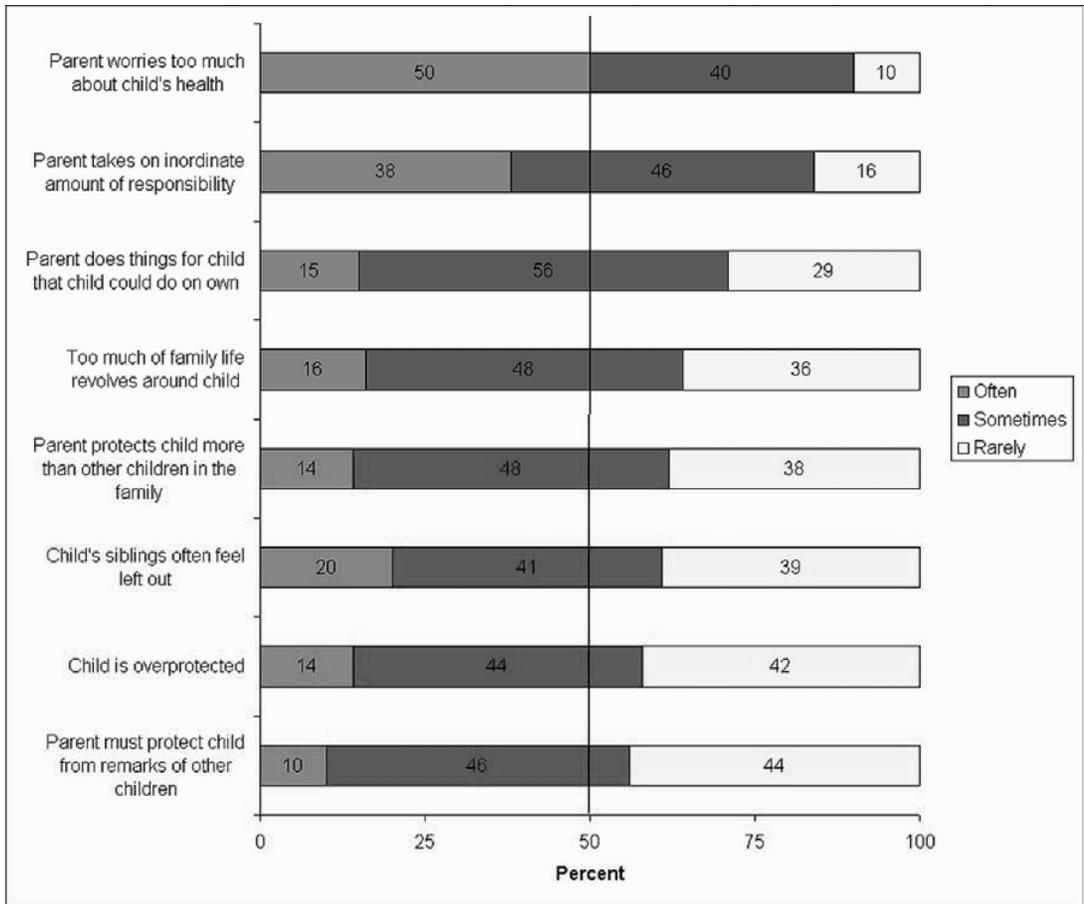


Figure 3. Percentage of Parents Responding “Often or Very True,” “Sometimes or Somewhat True,” or “Rarely or Not True” to Family Positives/Strengths on the Cystinosis Behavior Questionnaire.

opinion were noted between mothers and fathers (responses for primary and secondary caregivers were examined, mother  $n = 59$  and father  $n = 39$ ). Mothers reported (or were more willing to report) the negative impact cystinosis is having on their child and the family. Mothers, more often than fathers, reported that their child has very negative feelings about the illness, and they reported that they are negatively affected by the disease in terms of time, worry, and financial issues. More mothers than fathers have discussed the seriousness of the illness with the child, have had the child share feelings with her or him, and have shared her or his feelings with

the child or another adult. See Table 1 for a detailed summary of our findings on parental differences.

These differences of opinion between mothers and fathers, although striking, are not surprising and have been documented previously in studies of behavior in chronically ill children, namely that informants often differ in opinion (Lavigne & Faier-Routman, 1992; Perrin, Ayoub, & Willett, 1993). Moreover, studies have shown that mothers of chronically ill children often have higher levels of distress than fathers (Kazac, 1987; Perez, 1997; Perrin et al., 1993; Tavormina, Boll, Dunn, Luscomb, & Taylor, 1981), thought to be



*Figure 4.* Percentage of Parents Responding "Often or Very True," "Sometimes or Somewhat True," or "Rarely or Not True" to Family Negatives/Stressors on the Cystinosis Behavior Questionnaire.

related to the greater care demands placed on the mothers (Lamb, 1986; Russell, 1986). Previous research has shown that maternal distress can have an effect on the child's adjustment and coping (Barakat & Linney, 1992). This difference between mothers and fathers in viewing the impact of the illness on the child and the family can lead to increased stress and frustration. Anecdotally, in many of the cystinosis families we have seen for our research, fathers are often the providers for the family and mothers are the caregivers. Therefore, this difference of opinion may be a byproduct of the proximity of the parent to the child in daily life.

### Academic Difficulties in Cystinosis

In terms of school performance, some children with cystinosis have documented neurological findings that have been found to have an impact on academic achievement (Ballantyne et al., 1997). Although 57% of parents of children with cystinosis reported that their child rarely had academic difficulties for which she or he receives special help, 30% reported that their child often had academic difficulties for which she or he receives special help, and 13% responded that their child sometimes had academic difficulties that need special help. In our experience, children with cysti-

Table 1

*Differences in Responses Between Mothers and Fathers on the Cystinosis Behavior Questionnaire*

Question	Mothers endorsing (%)	Fathers endorsing (%)
<b>How do you think your child feels about having cystinosis?</b>		
• Hates it, unfair, unhappy	66	29
Examples from parents: Hates it; scared; said he wished he didn't have it; none of friends have to take meds; angry at taking meds and wetting bed; wonders why him and not sibling; diapers make him feel like a baby; doesn't like being short; doesn't like being different; hopes she doesn't die soon		
<b>How do you think your child feels about having cystinosis?</b>		
• OK, good, handles it well	34	41
Examples from parents: Doesn't mind; loves life; sometimes likes the attention; no big deal; handles it well; proud, like it's a club; doesn't feel different at all; accepts it as fact and gets on with life; remembers nothing else but cystinosis; doesn't feel any worse than having a cold		
<b>How has this illness affected your life?</b>		
• Time consuming, worried, financial issues	75	42
Examples from parents: Consumes our life; hurts deeply; life revolves around child, no more social life; financial, emotional, and physical burdens; it is exhausting. I need more support; can't hold a permanent job; everyday dealing with meds, lab work, having to be awake at odd hours; hard to think of ourselves; not as free to go places, take fewer vacations; worry is always there; sibs get less attention and feel less loved		
<b>Have you discussed the seriousness of this illness with your child?</b>		
• Yes	78	49
Examples from parents: Knows she needs to take meds so she doesn't get sick; discuss as much as possible; knows about the treatments and what they prevent; discuss illness with child in a positive way; knows he will have disease for his whole life; knows disease makes her feel sick; knows to tell me if he feels physically different in any way		
Are you able to speak of your feelings about your child's illness freely with your child?		
• Yes	78	49
Examples from parents: Knows I love him and I'm sad that he's sick; we talk when there are problems; yes, but I don't want to scare her; yes, but cautious not to really cry in front of him		
<b>Does your child ever talk with you about his or her illness?</b>		
• Yes	71	46
Examples from parents: Often says he hates cystinosis; child will ask for advice when teased; child talks about symptoms and treatments to stay healthy; asks if she has a question; child states that she is scared; asks if he has to take meds when he is grown up; asks questions about water intake and diapers at night; asks about meds and the blood tests		
<b>Does your child have any impairments from cystinosis that are noticeable to others?</b>		
• Yes	76	51
Examples from parents: Small size, looks younger than age; photophobia; tubes from chest and stomach; Cushingoid features; scars; takes a lot of medicine; thinner and paler; lack of strength, can't exert self; bad breath and body odor from medication; frequent urination; legs/knees look funny; poor fine motor coordination; speech and reading problems		
<b>Are you able to speak freely of your feelings about your child's illness with your spouse or a close, supportive adult?</b>		
• Yes	93	71
Examples from parents: Yes, very easily; yes, but not with spouse, don't share fear or negative feelings with him; yes, once I get started. . . sometimes I hold back; yes, but I wish for a better support system		
<b>How do you feel about your child's illness?</b>		
• Optimistic	66	46
Examples from parents: Believes there will be a treatment and a cure one day; optimistic about renal transplant; cautiously hopeful about medication and kidneys; hopeful; runs the gamut but remains optimistic; optimistic but feel it's not fair		

nosis may have real cognitive or academic limitations (e.g., nonverbal learning disability) that are sometimes not recognized in the school setting; instead, poor school performance may be misattributed to other factors such as laziness. On a more positive note, parents reported that behavior problems at school that cause the child to be sent to the principal or counselor seem to be much less of a problem (85% rarely, 6% sometimes, 0% often).

### **Interventions in Cystinosis**

Parents of children with cystinosis overwhelmingly endorsed some negative behavioral characteristics in their child, family adjustment issues/stressors, and academic difficulties. We thus find it quite striking that 80% of the children in our sample rarely see a therapist for social and/or emotional issues, whereas 10% sometimes see a therapist, and only 7% often see a therapist. This is consistent with previous research (Gortmaker, Walker, Weitzman, & Sobol, 1990; Smyth-Staruch, Breslau, Weitzman, & Gortmaker, 1984) as well as anecdotal discussions with parents of children with cystinosis that suggest that a majority of time is spent on the medical management of the disease, thus psychological treatment of the child and family may be ignored. In addition, in the school setting there may be a tendency for teachers to accept subtle learning difficulties as a product of the illness and not make referrals for intervention as readily for the chronically ill child (Baskin, Saylor, Furey, Finch, & Carek, 1983; Deasy-Spinetta, 1993; Sexson & Madan-Swain, 1995).

The literature indicates that when children with chronic illness exhibit behavior problems, there is a need for behavioral management, as well as other forms of services like respite care, after school and recreation programs, and summer camps (Floyd & Gallagher, 1997). Evidence suggests a beneficial effect of intervention programs for chronic disorders (e.g., psychological interven-

tions directed at disease-related or emotional/behavioral problems; Gortmaker et al., 1990; Kibby, Tyc, & Mulhern, 1998). These children may benefit from behavior therapy, including concrete practice, problem solving, and rehearsal in dealing with social situations. It is also important to teach adolescents how to be comfortable and assertive with peers while complying with their medical regimen (La Greca, 1990). Research findings have indicated that social support, both from other children with chronic illness and from typical children, may also help with self-esteem and compliance (Varni et al., 2000). Moreover, research on children with diseases that affect the central nervous system has indicated that consistent routines are especially important because they provide structure and clarify expectations (Loftin & Koehler, 1998). Routines may be a particularly vital target for intervention in cystinosis because establishing disease management routines (e.g., medication every 6 hrs, eye drops every 3 hrs, and proper hydration) as young children may help in the transition to adolescence, at which time these teenagers are expected to start independently managing their disease.

### **Narrative Information Provided by Parents**

Given the focus of this article, it is important to capture, in the parents' own words, some of their insights, perspectives, and issues. Table 2 presents written narrative information provided by parents regarding how cystinosis has affected their lives, their children's temperament, a description of temper tantrums in their children with cystinosis, and the need for intervention. The themes in the narratives depict the immense impact of cystinosis on the children's and families' lives, the struggle to maintain a sense of normalcy in their daily lives, and the extremes of temperament and intensity of temper tantrums in their children with cystinosis. Information such as this can lead to a greater under-

Table 2

*Written Narrative Descriptions Provided by Parents of Children With Cystinosis (Identifying Information Has Been Deleted and Names Have Been Changed for Purposes of Confidentiality).*

Parent	Narrative
<b>Parents' responses to the question "How has this illness affected your life?"</b>	
Mother A	"It has changed our life <i>greatly</i> —more than I can say. There was life before Robert's illness, and life after. There is not a day I live I don't think about cystinosis or its impact on my son. I can talk about it intellectually. I get unexpectedly teary discussing the emotional aspects. When he is well I focus on the day to day and try to enjoy every minute with him. When he is ill, the fragility of his health makes me sad and reminds me of the unthinkable, unbearable reality that we could lose him. Through all the pain, though, I will always see Robert's life as a gift to us. His joy, enthusiasm, and courage is something I treasure. The challenges and opportunities for growth we would never have experienced without him are immense." (6-year-old boy)
Mother B	"All day long he is what I'm thinking about. I get very stressed out sometimes, to the point where I just what to cry all day long, but I would never change the fact that I have him because I love him more than anything in the world. I just wish he didn't have this disease." (3-year-old boy)
Mother C	"Ethan's life has been one shock after another affecting us in different ways. We have not slept through the night in 5 years, we don't go anywhere without water and knowing where the next bathroom is. I always have chips in my purse. I feel worried and/or stressed most of the time. Ethan has learning problems, i.e., cueing and auditory memory problem which requires attention and consideration at home and at school. It has affected all of our eating habits in that we have almost no rules concerning when and where we eat. It has affected us economically due to extra costs and my inability to work full time because of the number of appointments, Ethan's sick time, and my own lack of sleep. There's been a lot of strain on my marriage and on our extended family and friends. Hospital stays are always added stress. Time and effort to prepare medications, cook different food for him, and carrying him when he's tired. We intend on buying a stroller for him soon to save our backs! Lots of negatives for such a positive kid!" (5-year-old boy)
Father D	"It has affected every aspect [of my life]. I don't have a social life of my own. I go out with my wife alone once every 4 months for a movie and dinner. We have [no] sex life. . . I'm not a workaholic or 'climber' at work. I try to enjoy and savor each day." (6-year-old-boy)
Mother E	"We still try to do all the family vacations but my husband and I are not able to go away like we used to. Claire needs someone who can handle her meds, feedings, etc. We go on separate holidays now. Vacations are fewer also because a lot of her meds are not covered and cost A LOT of money." (6-year-old girl)
Mother F	"Now that it's 8 months since her diagnosis, life is getting 'more normal' again. Medication [giving it] has become less of an issue, although there are days when I feel like that's all I do. Life has been broken down to 6-hour increments. It also means I need to be even more organized when it comes to planning my day; taking meds and water with wherever we go, etc. My husband and I have less alone time together because we keep Sabrina up until we go to bed to give meds. Even when we go out by ourselves, we have to consider the time and whether our caregiver can give meds. The responsibility of the medication, and therefore keeping Sabrina healthy, could be totally overwhelming if I let it be." "Because she is doing so well—looks (except for being petite), and acts like any other 2-year-old, it would also be easy to live in denial as well. I've had a couple occasions where we're out doing something fun, enjoying the moment, and I've forgotten that there was anything different about our lives. Then I looked at my watch, remembered the medication schedule, and came crashing back to reality! The positive changes are becoming more evident as we go. The first being that I love and appreciate Sabrina even more! I thoroughly appreciate and find joy in all the 'normal kid' things she does. Watching her splash in rain puddles the other day suddenly seemed like it was the most important thing I could be doing. I don't take things like these for granted. Sabrina's condition has also brought our family together as more of a team. We work together more. Our faith and dependence on God has also grown." (2-year-old girl)

*(table continues)*

Table 2  
(continued)

Parent	Narrative
<b>Parents' responses to the question "How has this illness affected your life?"</b>	
Mother G	"Cystinosis affects our lives in every day we live. . .mixing medications, doing lab work, talking to parents, getting up at odd hours to give meds, or help Tara to the bathroom. Yes, our lives have changed forever by this! At least as Tara has gotten older, it's become easier for us." (7-year-old girl)
Mother H	"Who's to say what my life would be without it. We've had it for almost 13 years and its just a part of life. It takes more time making up meds and administering every day—and there is no question you worry. But you worry about all your children, with or without cystinosis. I sometimes get very emotional <i>after</i> clinic appointments or hospital stays. Nothing has to happen—it's just the normal stress. It's sometimes frustrating the limited medical knowledge I have. I've learned <i>a lot</i> but there is so much I don't know, and I fear that Thomas may be suffering from my lack of knowledge. I know he has excellent medical doctors caring for him but I feel if I knew more he'd have all the more going for him. Thomas can be the sweetest but of late (could be his age) he is hard to get along with. I hate to give in to him just so life is bearable—so I haven't and it isn't!" (14-year-old boy)
Father I	"We have learned that centering a family around a child with a special need is COUNTERPRODUCTIVE. Everyone is just as important as the next. Cystinosis has not slowed us or Christine down. In restaurants we are able to pour meds down the G-tube with one hand and eat with the other. . . . If she does NOT receive a whole lot of special attention <i>now</i> , she won't expect it from society the rest of her life. Christine LEADS A NORMAL LIFE!!!" (4-year-old girl)
Mother J	"Cody's illness has made a huge impact on my life. Initially, when Cody was diagnosed almost 4 years ago, I had to completely change my 'plan' of what I wanted in life. I had to adjust to the transition [from] being a full-time working mom, contributing half to the family's income, to a stay-at-home mom not able to contribute anything financially to our family. Initially, I really missed the adult contact with coworkers and the freedoms that working outside the home provide. After an adjustment period of a few months, I decided to focus my life on the positive priority of trying my best to keep my family healthy and happy. Instead of dwelling on any negative aspects associated with a chronic disease, I tried to help our family live a life of happy and meaningful moments. Simultaneously I am trying to find the best medical care for my son. I refuse to listen to any professional that states "they can't do anything more to help Cody." I try to keep my children in the company of positive and happy people. My son's illness has taught me the true value of living a happy, fulfilling, productive life. [Cody's sibling] and Cody have coped with cystinosis with huge amounts of love, caring, honesty, courage, and bravery. I have learned how to prioritize the important aspects of living from my husband and children because of the positive way they have dealt with cystinosis." (4-year-old boy)
Father K	"While there are many negative aspects and pressures associated with being the parent of a chronically ill child, I believe it has provided me with a more balanced perspective on life and has given me an incentive to better balance the needs of my family and career pressures." (4-year-old boy)
<b>Parents' descriptions of temperament in their child with cystinosis</b>	
Mother L	"By 3 she was very defiant and strong willed—had violent temper tantrums. . . . We all have tempers but Olivia's seems to be the most intense; she also is the 'most' in all <i>behaviors</i> —guilt, happiness, etc." (9-year-old girl)
Mother M	"He's a Jekyll/Hyde-either as 'sweet as he can be' or a 'holy terror.' He's stubborn. The extremes in mood and <i>quick</i> changes seem to be more intense [than other children]." (6-year-old boy).
Mother N	"I attribute some of Marian's behavior and traits to a 'difficult temperament' (versus easygoing). She has been stubborn and strong-willed since newborn. I also think that some of her negative behaviors are due to being diagnosed at age 2 years—so she was ill and sickly and <i>PAMPERED</i> at an age when others were 'setting limits.' (I particularly was <i>not</i> willing to set limits to discipline Marian until a much later time!)" (7-year-old girl)
<b>Parents' descriptions of temper tantrums in their child with cystinosis</b>	
Mother M	"Cole is my challenge in life. One minute he is loving and the next you don't want to be near him. . . . [The intensity is] especially more significant compared to other children. . . . I guess my main concern with Cole is his behavior. I sometimes feel like a 'first-time' mom [although I am not]. I really don't think I've changed my method of rearing—if anything, I would hope it has gotten better. I feel my hands are tied because nothing seems to work." (6-year-old boy)

Table 2  
(continued)

Parent	Narrative
<b>Parents' responses to the question "How has this illness affected your life?"</b>	
Mother N	"It [the intensity of tantrums in her child with cystinosis] is more intense than her [siblings]—It's louder, more extreme, and lasts longer. Marian cannot be redirected easily when upset—she just had to run her course!! (Ignoring some/most of her 'raging' behavior seems to help!)" (7-year-old girl)
Mother A	"Robert throws great tantrums if he doesn't get his way. . .He hollers, will call us names (usually Dumbo), will hit out at us. Not real hard but hard enough. . . .Our first child <i>never</i> had tantrums like this." (6-year-old boy)
Mother O	"He is usually such a happy child. I can see 'it' coming now—when he is tired or doesn't feel well he is more likely to begin crying and screaming. He is much more aggressive now than he used to be, i.e., hitting, kicking his younger sister. Many times it will happen after a nap—he'll begin screaming and yelling for no apparent reason. He doesn't want anyone to touch him and he tells us to go and when we do he cries telling us to come back. He also does it after he's been told 'no.' He'll suddenly go from good mood to very mad. . . . His father and I try to talk very calmly to him but after awhile one of us will 'give up' and leave him to the other to deal with." (3-year-old boy)
<b>Parents' descriptions of need for intervention</b>	
Mother P	"His behavior has really been bad lately. Hopefully it's just a 'phase.' I said when he was first diagnosed I needed to take a class in how not to spoil rotten a sick child. Nothing was available to us and now I think we created a monster." (6-year-old male)
Mother N	"Marian also has begun receiving special ed this past school year—she displays difficulty with fine motor coordination and academics (math and reading). Since receiving 'extra' one-to-one each school day, Marian's attitude has improved (in general). I know Marian 'knew things were too hard for her' and she was relieved to receive extra help. Additionally Marian's behavior was 'being shaped' at school via behavior management/reinforcement—and we also made a point of carrying more 'behavior shaping' at home. I am not all that sure that Marian would have had learning problems without cystinosis. . . . I feel like we've made good progress on Marian's behavior since 1 year ago. Getting concentrated help and feedback via our school has been helpful and has forced us to really focus our concern on behaviors <i>we wanted to see changed!</i> " (7-year-old girl)

standing of the biopsychosocial demands of cystinosis and has the advantage of depicting parents' unique experiences rather than group trends.

### STRENGTHS AND LIMITATIONS OF THIS STUDY

We often hear questions such as the following from already overburdened parents caring for a child with cystinosis: "What am I doing wrong?" "Why is *my* child so difficult?" and "Am I alone, or does anyone else have these problems?" This article documents the complexities (both strengths and difficulties) that families with a child in the chronic stage of cystinosis face every day.

This information can serve as a valuable resource for parents to read and share with school personnel and physicians working with children who have cystinosis.

One limitation of the current study is that a comparison group was not included. The results, therefore, are an illustration of the issues faced by families dealing with cystinosis. We are not implying that children with cystinosis have more (or fewer) behavioral difficulties than children with other chronic disorders or typically developing children. We merely hope to identify and illustrate areas of strength and weakness for these families, regardless of how the frequency of these issues compares

with another group of families. In future studies, it will be interesting to see how these cystinosis results compare with those for other chronic illnesses. Moreover, although the particular strengths and weaknesses documented in this article relate specifically to cystinosis, the findings presented here may be a resource for families with children who have other chronic medical conditions and who may also be experiencing behavioral and adjustment issues. It should also be noted that the data were collected from parents or guardians; future studies should include views of the children themselves through a self-report questionnaire.

Another potential limitation of the current study was that we used a questionnaire developed in our laboratory rather than a standardized, published questionnaire. Although the lack of normative data does not allow for normative comparisons, such an analysis was not the purpose of this article. Quantitative results comparing behaviors in children with cystinosis, cystic fibrosis, and controls have been published previously by our laboratory (Delgado et al., 2005). This study aimed to go beyond the numbers and allow for a greater understanding of the qualitative experiences of families affected by this disease.

One of the strengths of this study is that we have been able to collect qualitative behavioral data in a homogeneous group of 63 children and families affected by cystinosis, which comprised approximately 20% of all individuals (both adults and children) in the United States with the disorder (Elenberg, 2003). Well over 90% of individuals who were given the questionnaire returned it to us, thus reducing the concern of a biased sample. A potential criticism of the current study is that the information was gathered via a questionnaire rather than an interview. Owing to the fact that cystinosis is an extremely rare disorder and these families live all over the United States (and around the world), and there is

very little funding available for research on this orphan disease, it was neither practical nor feasible to travel around the country to conduct interviews.

There is a paucity of information in the literature describing the biopsychosocial demands of cystinosis; hence, this initial presentation is critical in terms of documenting potential adjustment issues within the child and family system. In fact, one of the strengths of this article is that we were able to collect detailed data on behavior and adjustment in a large percentage of families affected by cystinosis. Many studies on behavior in chronic illness group children with various chronic conditions together, thus any disease-specific correlates are lost. Given that cystinosis has been documented to affect the central nervous system, it is likely to have specific effects on cognitive-behavioral functioning beyond the general or common effects of chronic illness. We believe that the presentation of descriptive data is a vital step in the process of better defining the complexities of these children and families.

## CONCLUSIONS

Cystinosis is a genetic metabolic disorder that is known to affect the central nervous system. Although the disease may affect particular areas of cognitive functioning, the effect on behavioral functioning is less well defined. The present study examined behavior and adjustment in a large sample of children with cystinosis and their families. We found that parents reported many areas of strength within their child and their family along with many areas of difficulty or stress. It is interesting, however, that very few of the families had sought out intervention for behavioral or adjustment issues, which warrants further investigation in future studies. Our finding of difficulties and stress in families of children with cystinosis highlights the need for health providers or others to closely monitor the psychosocial course of the child and family.

The fact that these children and families demonstrate numerous strengths, despite their difficulties, is a particular asset for formulating interventions. Namely, child and family strengths can be productively used in formulating appropriate and effective interventions, and in helping parents and children cope with the challenging situations they face. In better defining behavioral and adjustment issues in cystinosis families, the current findings may prompt greater awareness in individuals caring for or working with a child with cystinosis. This awareness is a critical element in helping to optimize the child's behavioral functioning and family adjustment.

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