

Psychosocial Aspects of Patients With Niemann-Pick Disease, Type B

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Health-care providers have only begun to understand the medical aspects of Niemann–Pick disease type B (NPDB), a relatively rare disease. Even less information is known about the psychological effects of living with NPDB. Patients with NPDB and their families face numerous psychological stressors including extensive medical testing, uncertainty of diagnosis, living and coping with a chronic illness, and grief and bereavement surrounding this progressively debilitating, and, ultimately, fatal disease. We used a qualitative case study approach to explore the human experiences of NPDB patients and families. To assess psychosocial adjustment, all participants were administered a semi-structured, qualitative interview, as well as quantitative measures. Five major findings emerged: (1) limited physical activity, social isolation, and peer rejection were identified as significant stressors; (2) stressors had a specific impact during the age span of 10–16 years; (3) parents and adult patients expressed frustration regarding the lack of available information and treatment; (4) patients described close family relationships as a way of coping with illness; and (5) adult patients identified early medical experiences as having a considerable psychological impact. The results of this investigation highlight and expand awareness of the psychological and social needs of NPDB patients and families. This study calls for a collaborative, multidisciplinary effort in the treatment of these patients and their families.

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INTRODUCTION

Niemann–Pick disease (NPD) belongs to the group of inherited lysosomal storage disorders (LSD) and is considered to be a rare, chronic, progressively debilitating, and ultimately, fatal disease [Minai et al., 2000]. An inborn error of metabolism that is inherited in an autosomal recessive manner, NPDB is caused by the deficiency of activity of acid sphingomyelinase (ASM) and the subsequent accumulation of sphingomyelin, cholesterol, and other lipids within cells and tissues of affected individuals [Schuchman and Desnick, 2001]. There are two different phenotypes of ASM deficiency NPD,

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types A and B, classified by severity, age of onset, and central nervous system involvement. NPD type A is a severe neurologic disease that leads to death by 3 or 4 years of age [McGovern et al., 2008]. In contrast, type B individuals generally have little or no neurologic involvement and may survive into late childhood or adulthood. Type B disease is characterized by hepatosplenomegaly, thrombocytopenia, interstitial lung disease and dyslipidemia [McGovern et al., 2008]. To date, there are approximately 1,200 cases of NPD types A and B worldwide, with the majority being type B (NPDB) [Schuchman and Desnick, 2001; www.nnpdf.org].

This study focuses on patients with NPDB. To live with NPDB is to exist with a chronic, debilitating, painful disorder. Patients with NPDB may not develop all the possible symptoms associated with type B, and the spectrum of severity is broad. Nonetheless, the signs and symptoms can be extensive, life altering, painful, and ultimately, life threatening. Until a diagnosis is made, NPDB patients may be subjected to ongoing diagnostic examinations and procedures. For children and adolescents, NPDB impacts psychosocial development. In addition, families are affected by the financial costs,

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uncertainty of diagnosis, and ultimately, grief and bereavement surrounding the loss of their loved one.

The clinical presentations of heritable metabolic disorders are variable, and the physical and psychological concerns are likely to be quite unique for individual disorders [Shapiro, 2002]. Research investigating the psychological issues for patients with distinct inborn errors, including maple syrup urine disease (MSUD), Fabry disease, and Gaucher disease, is starting to emerge [Packman et al., 2006, 2007]. At this time, no studies exist that examine the psychological impact of NPDB.

Given the limited research on patients with NPDB, it is vital to gain greater understanding of how this disease affects the psychosocial functioning of patients and their families. Specific knowledge of common features of NPDB patients can aid health-care providers in devising comprehensive treatment plans: that is, plans addressing both physical *and* psychological concerns. The study's goals were to assess the psychosocial adjustment of patients as well as to enhance our understanding, sensitivity, and awareness of the rich human experiences of those who live with a chronic illness. From this understanding, health-care providers will be better able to guide their patients with inborn errors of metabolism.

MATERIALS AND METHODS

Overview of Data Analysis

A qualitative case study methodology was used to explore the human experiences of NPDB patients and families. Yin [1993] describes the case study as an examination of phenomena in a real-life context when the boundaries between the phenomena and the context are not clearly defined and multiple sources of evidence are used. A case study allows an investigation to retain the holistic and meaningful characteristics of real-life events.

In the present study, the experiences of psychosocial development and adjustment occurred within the real-life context of patients and parents living with a chronic illness. Therefore, the case study method facilitated the identification of specific factors affecting these patients and families and their unique responses to them. Interviews with adolescents and their parents and adult patients elicited information regarding life experiences, development and school, relationships, and medical experiences in the context of NPDB. All of the interviews were coded by the first author and half were coded by the second author. Discrepancies in coding were discussed and reconciled between the two coders. Themes were identified and illustrative quotes were selected to explicate our findings.

This study also used quantitative methodologies to supplement the qualitative findings. Specifically, we used the Measures of Psychosocial Development (MPD) [Hawley, 1988], a standardized assessment tool based on the developmental theory of Erikson [1968].

Participants and Procedures

Participants were patients with NPDB between the ages of 13–18 (adolescent group) and their parents, and 19 and older (adult group). The total sample was 17 participants. A study announcement was published in the National Niemann-Pick Disease Foun-

dation Newsletter. All of the study participants were interviewed at the annual Niemann-Pick Disease Family Conference [August, 2005]. All procedures and materials were approved by the Institutional Review Board (IRB) at Pacific Graduate School of Psychology.

A questionnaire designed for this study was used to gather background information on the participants. The items were designed to obtain current demographic data, information on medical, psychological, and education history, and for patients, disease symptoms.

Psychosocial adjustment was conceptualized as the presence or absence of psychological or emotional symptoms that are experienced to a degree that cause discomfort, impaired daily functioning, and interpersonal difficulties. For adolescents in particular, positive psychosocial adjustment can be reflected in the presence of few behavioral problems, age-appropriate functioning in school, with family, and peers, and increased quality of life.

To better understand the experience of patients and families, semi-structured interviews were designed by the investigator to gather information about the psychosocial impact of NPDB. The patient interview consisted of seven domains: (1) Life experiences with respect to NPDB; (2) feelings about having NPDB; (3) relationships with family and friends; (4) developmental history and schooling; (5) college; (6) employment; and (7) medical experiences. The parent interview included the same domains except developmental history and schooling. The interview items were drawn from several sources [Arnett and Taber, 1994; Tong et al., 1998; Hartman et al., 2000; Palmer and Boisen, 2002; Mellin et al., 2004; Packman et al., 2006, 2007]. The items and content areas for the interviews were reviewed by a panel of experts consisting of a geneticist, two genetic counselors, members of the NNPDB board, and a pediatric psychologist and their suggestions were incorporated into the final versions.

RESULTS

Description of Sample

A total of 19 families, including adult patients and child/adolescent patients with their parents, expressed interest in the study. Of these, 8 were excluded for the following reasons: patient was not over 13 years of age ($n = 2$); patient was NPD type A or C ($n = 4$); family or patient was not available to complete the study ($n = 2$). The final sample of 17 participants included 4 adults, 4 adolescents, and 9 parents. Four of the nine parents were related to the adolescents in the study.

Demographic data and background information for the whole sample ($n = 17$) is presented in Tables I and II.

Qualitative Data

The results of the semi-structured interviews with patients and parents are presented with a focus on five different themes corresponding to interview categories. The remainder of the results section reports how participants perceived each of the interview categories. Supporting quotations for each theme are listed in Table III.

TABLE I. Demographic and Background Information—Adolescent and Adult NPDB Patients

Variable	Patients (n = 8)
Gender	
Males	3
Females	5
Current age	
Mean	28.4 years
Range	16–43 years
Age of diagnosis	
Mean	7.9 years
Range	1.5–37 years
Highest level of education	
11th grade or less	3
High school diploma or equivalent	3
1–3 years of college or equivalent	0
4 years of college or more	2
Ethnicity	
Caucasian	8
Religion	
Catholic	3
Jewish	0
Protestant	0
Other: Christian, Methodist, Baptist	5
Medical history	
Arthritis	1
Chronic lung disease	3
Chronic allergies	1
Trouble seeing	2
Back problems	2

TABLE II. Demographic and Background Information—Parents of NPDB Adolescents

Variable	Parents (n = 9)
Gender	
Males	4
Females	5
Current age	
Mean	45.4 years
Range	38–53 years
Highest level of education	
11th grade or less	0
High school diploma or equivalent	4
1–3 years of college or equivalent	5
4 years of college or more	0
2 or more years of graduate school	0
Marital status	
Married/partner	5
Divorced	4
Single	0
Ethnicity	
Caucasian	9
Religion	
Catholic	4
Protestant	1
Other: Christian, Methodist, Baptist	4
Total yearly income	
Less than \$25,000	0
\$25,000–\$50,000	2
\$50,000–\$75,000	4
\$75,000–\$100,000	1
More than \$100,000	2

Limited physical activity, social isolation, and peer rejection. The study's findings were consistent with past research on the psychosocial adjustment of patients and families with a chronic illness [Packman et al., 2006, 2007]. The majority of participants reported limited physical activity as a result of both fatigue and the life-threatening danger posed by some activities. This finding is consistent with McGovern et al. [2008] who reported that adults with NPDB do not consider themselves to be as healthy as others and expect their health to worsen over time. The participants in our study also described peer rejection and social isolation.

In addition, patients in the present study reported that NPDB had a significant impact on their daily lives. Two of the adult participants felt particularly psychologically affected by their illness, and noted that having NPDB had significantly affected their emotional stability, personality, and ability to fulfill traditional gender roles. This finding echoes that of Ware [1999] who described role constriction among individuals with a different chronic disorder, chronic fatigue syndrome. In the present study, participants reported a decreased ability to fulfill role demands and participate in school, employment, and relationships.

Close family relationships as a way of coping. One parent highlighted themes of worrying about the progression of the disease, being overprotective, and balancing the needs of his

daughter and her siblings. Another adolescent's father relayed that his son first indicated awareness that something was wrong when he was 6 years old. For this father, this was the moment he realized that trying to protect his son by ignoring or avoiding the topic was no longer working. From that point on, he described always answering his son's questions to the best of his ability.

Impact of NPDB on psychosocial development. Researchers have identified additional areas of concern for children and adolescents with chronic illness including self-esteem, school functioning [Packman et al., 2004; Storch et al., 2008] and quality of life [McGovern et al., 2008]. We found a discrepancy in parent and adolescent perspectives regarding the most difficult age in terms of coping with NPDB. In the current study, parents rated high school as the most difficult age and time of life for their child with NPDB. It appeared that physical growth issues and social isolation were most distressing to parents. The patient interviews indicate that high school was indeed a challenging time for exactly the reasons parents identified: difficulty making friends, dating, and participating in social events. In contrast to parents, patients identified elementary school as equally challenging as high school. Adolescent and adult patients recounted memories when their grade-school peers treated them differently and with cruelty.

TABLE III. Supporting Quotations for Lived Experience of NPDB

Interview Category: Theme

Life Experiences with Respect to NPDB:

Limited physical activity, social isolation and peer rejection

"I couldn't play the games that the other kids were playing. And then I used to get made fun of, called pregnant, because of the way I used to lean back to hold the extra weight of my stomach. I've kind of trained myself now how to walk. I remember I used to come home crying. The kids don't know that it hurts you that bad."—39-year-old woman

"I would love to be able to go hiking, do more traveling. I just don't have the capability to do that. I think that my personal life might be different as well. I don't have the energy to dedicate to dating and finding someone to be a partner in my life, because all of my energy right now is spent just getting from day to day."—39-year old-woman

"I had some friends at church. But I'm not able to run around and do all the stuff that they do. It kind of leaves you out of the loop."—43-year-old woman

Relationships with Family and Friends:

Coping within the family

"We were driving and my son spoke up and said, 'Daddy, am I gonna die?' And I said, 'well yeah, of course you're gonna die,' 'I'm gonna die, everybody dies.' And he said, 'no, I mean I'm sick aren't I.' And I said, 'yes, we found out that you were and we really don't know what to expect.' And he said, 'it's ok, I feel ok.' And that's when I realized we hadn't actually told him and that what I was doing was adversely affecting him, because he worried about me. And a kid that's six shouldn't have to worry about mom or dad."—Parent of 18-year-old young man

"Me and [my brother] can relate really well about it. And [my other brother], he knows all about it, but he doesn't have it so he doesn't know what it's really like. He's okay about it, but some issues he doesn't really understand."—18-year-old young man

"It's caused considerable tension. It's the 600-pound gorilla in the room. It causes a lot of tension between us because we have different opinions on what my son is capable of."—Parent of 18-year-old young man

Developmental History and Schooling:

Impact of NPDB on psychosocial development

"As a child I had an enlarged spleen and liver. Whether my parents were overprotective or not, they just didn't want me to get hurt. So I couldn't play soccer, wrestle, rough-house. Throughout my childhood I heard, 'you can't, you can't, you can't.' And the fact that everybody else was while I stood by caused me to be fairly shy, not outgoing, to be lacking in self-confidence, possibly to be an underachiever in scholastics and occupationally."—41-year-old man

"We've had him on growth hormones for a number of years and he's made a dramatic increase in height. He was in high school and he wasn't even 4 feet tall. And so it was a huge thing, especially when the other guys start getting interested in girls, hanging out with somebody who looks like your little brother doesn't work very well. So, he's had problems with friends in that respect. Then when he started getting interested in girls, that was a huge problem. Because a 16 year old girl doesn't want to go out with somebody who looks 12. So that part was tough. How do you explain how to deal with something like that? And there's no street light, you're walking in the dark. I don't know what to tell him."—Parent of 18-year-old young man

"Compared to other kids her age, her body is just not maturing and she doesn't act her age either. Other kids are developing and changing and she's staying status quo and not reaching puberty."—Parent of 16-year-old young woman

Medical Experiences:

Frustration regarding lack of information and treatment

"At first it was scary, not knowing what we were looking at. There was so little information and we got a lot of conflicting information from doctors. Immediately after diagnosis there was a lot of grief."—Parent of 18-year-old young man

"It was a relief to know that something was wrong. But then it was disappointing to know that there was nothing that they could do about it."—43-year-old woman

"I understand more what the doctors are saying. I wish they'd just tell me. Sometimes I overhear, like when they measure my stomach or liver and spleen. I worry they'll get really big. Sometimes I overhear them say I need to get an MRI and I don't like those."—16-year-old young woman

Psychosocial impact of medical experiences

"As a kid I got poked and prodded by the best of them. I had a liver biopsy, a spinal tap, had my liver and spleen measured. When I was in the hospital, every intern had to poke at me. Because you weren't gonna see one of me. I went to an endocrinologist every 6 months. He was doing his best, but one of his things that he did in these 6-month exams was measure my genitals. Therapy helped me put it together—well no wonder I'm insecure! It's things like that that you really didn't think of, or your parents didn't think would have a detrimental effect on you later in life. Maybe some people don't think twice about it."—41-year-old man

Parents' lack of awareness that their child first experiences distress before high school has implications for their ability to provide support and intervene. For example, one adolescent patient remembered that at age 10 years she was called pregnant because of the way she walked to support her enlarged organs. She described this as very wounding and reported that she would go home and cry every day. When this patient's mother was asked to recall her daughter's experiences in elementary school, she believed that her daughter was picked on, but stated, "I don't even remember what they would have called her. It wasn't anything big." While there are various reasons why this parent may not remember her child's early experience, the discrepancy between parent and teen report suggests that NPDB parents may need to increase awareness of their child's emotional well-being earlier than expected. It is not uncommon to find differences between parent and child reports as shown in studies of children with glycogen storage disease type 1, asthma, and cancer [Varni et al., 2002; Packman et al., 2007; Storch et al., 2008]. One possible explanation for the discrepancy found in the current study is that younger children are developmentally less articulate regarding psychological distress. Thus, parents' awareness of emotional upset may have increased with their child's ability to express distress.

Impact of NPDB on psychosocial development: Quantitative findings from the measure of psychosocial development. On the MPD, six participants—three adults and three adolescents—were assessed for resolution of Eriksonian stages. A low-resolution score suggests developmental stress resulting from lack of adequate resolution to a specific stage conflict [Hawley, 1988]. Though this is too small a sample size for statistical comparisons, it is of interest to note that half of the participants reported unresolved developmental crises at the stage of initiative versus guilt (ages 3–5 years). According to Hawley [1988], "there is a constant fear of making mistakes, being weak or inadequate. Overwhelming guilt is manifested by self-restriction, over conscientiousness, acquiescence, and paralysis of action." Individuals with high initiative are said to,

have a strong sense of purpose and a clear vision of what they want in life. Action, competition, adventure, and experimentation are their key words. They display a strong sense of motivation. High scorers are characterized by honest ambition, energetic drive in pursuit of accomplishment, a tendency to solve problems by planning and attacking, adventuresomeness, and a tireless "go-getting quality" [Hawley, 1988].

In addition, and consistent with the qualitative data, half of the participants expressed resolution conflicts in intimacy and isolation (young adulthood; 18–35 years). Finally, half of the participants expressed resolution conflicts in ego integrity and despair (late adulthood: 55–65 years), suggesting that there is some distress as a result of unresolved developmental conflicts. This domain appears to capture the existential perspective that many of the participants expressed: that they have missed opportunities and will never have a chance to experience certain aspects of life. This is especially true in the area of relationships. The MPD findings corroborate and complement interview data that showed that most participants

considered NPDB to have considerably impacted their psychosocial lives.

Frustration regarding lack of treatment and information.

Parents also had an opportunity to describe their concerns and challenges with regard to raising a child with NPDB. Parents reported feeling extremely frustrated around the paucity of medical and psychological knowledge and inadequate resources for treatment for NPDB. They spoke about how this lack of information impacted their experience emotionally when their child was first diagnosed.

Psychosocial impact of medical experiences. Patients reported feeling supported by their parents, especially with regard to medical experiences. One patient remembered his parents intervening on his behalf and refusing to allow any more tests, stating that he had been through enough. In a study exploring how adolescents with and without chronic illness perceive patient–physician trust, Klostermann et al. [2005] found that chronically ill children preferred to have their parents involved in their care. The authors identified specific behaviors to improve trust including asking for the adolescent's opinion and not withholding information.

This point was made clear by one participant as she described her medical experiences. As a 16-year-old who interacts regularly with medical personnel, this female adolescent patient has an adult level of comprehension of her medical status. She indicated that she is as familiar with the procedures and medical language as her parents, yet she lamented the fact that her doctor does not speak directly to her: "I wish they'd just tell me." Cognitively, this patient is mature enough to understand the implications of her illness and to experience the existential angst of mortality.

DISCUSSION

The present study examined the psychological aspects of patients and families with NPDB. The available research on chronic illness suggests that there are both positive [Mellin et al., 2004; Packman et al., 2005] and negative [Eiser, 1990; Holmes and Deb, 2003; Packman et al., 2004] psychological effects on individuals with various diseases. While commonalities exist in the experiences of individuals with chronic illnesses, how one copes with chronic disease is strongly influenced by the context in which the illness occurs: that is, family, peer group, school, work place, and developmental stage.

Similar to others living with chronic illness, the families in this study have a number of needs. However, what is evident from the findings, and because of a qualitative case study methodology, is that each family has their own unique set of circumstances that helps or hinders them in various areas of their lives. Whereas some families face extreme financial burden, others state they have a more difficult time with maintaining their emotional and mental health.

Five major findings emerged from the interviews: (a) all participants identified limited physical activity, social isolation, and peer rejection as significant stressors; (b) these stressors were especially notable for participants during the age span of 10–16 years; (c) parents and adult patients expressed significant frustration regarding the paucity of available information and treatment;

(d) patients described close family relationships as a way of coping with the illness; and (e) adult patients identified early medical experiences as having a considerable psychological impact.

The quantitative data were collected to corroborate the qualitative findings. Psychosocial adjustment, as it applies to the concept of transition to adulthood, was explored using Erikson's [1968] theory of psychosocial development. Psychosocial development was formally assessed in all patients using the quantitative Measure of Psychosocial Development [MPD; Hawley, 1988] which is based on Eriksonian theory. According to Erikson [1968], the crisis of trust versus mistrust characterizes infancy and the infant's need for a predictable and relatively consistent form of interaction with others. In the play age, children learn varying degrees of self-control through language acquisition, motor coordination, and more sophisticated interpersonal relationships. The issue is between becoming an autonomous, creative individual and an inhibited individual filled with self-doubt. By the age of 5 years, the child builds on his or her autonomy to engage in more self-initiated activities that build a sense of self-esteem. The psychosocial conflict is the development of a sense of initiative versus a sense of guilt. During the school years, developing cognitive capacities enable the child to broaden the basis of personal and social competence through advanced learning [Muuss, 1988]. The task is the development of a sense of industry versus the emergence of feelings of inferiority. With adolescence, there is the need to establish an identity unique to oneself and apart from the earlier identifications acquired during the formative years. The psychosocial conflict is a sense of identity versus identity confusion. During young adulthood (18–35 years) individuals seek mutually satisfying relationships; and the psychosocial conflict is a sense of intimacy versus isolation. With middle adulthood (ages 35–60), the task is producing something that contributes to the betterment of society (generativity) versus stagnation. Finally, in late adulthood, individuals may feel fulfilled with the sense that life has meaning (integrity). On the other hand, some adults reach this stage and feel despair [Erikson, 1968].

Interview data from the case studies indicate that the low-resolution scores may not be so much a conflict of this stage, but a manifestation of the chronic fatigue associated with NPDB. Thus, the "guilt" of this stage conflict may not be experienced as psychological distress. Rather, self-restriction, overconscientiousness, and lack of action appear to be self-protective factors adopted in the management of NPDB.

Based on previous studies and the findings of the current study, it appears that there is a discrepancy between what physicians' knowledge and patients' and parents' expectations for information and treatment. In the area of inborn errors of metabolism, Enns and Packman [2002] point to the importance of educating the patient and family and working closely with a multidisciplinary team. This will serve to distribute the families' care among health-care providers and provide realistic expectations for treatment and outcome. Research on coping and sense of control among those with chronic illness [Ell et al., 1989] suggests that adolescent patients may benefit from a more collaborative relationship among family and health-care providers, thereby providing them with a more balanced sense of control (i.e., assertive and yielding modes of control) and promoting better psychosocial health overall.

Increasing families' support from multiple sources is an important way of bolstering parents' coping skills. Two of the parents in the current study indicated that they seek support from friends and NPDB support groups as a way of coping with frustration regarding the lack of information and treatment. For example, one parent remarked, "the NPDB support group is very helpful because it's familiar. I know now that I don't have to invent a solution to everything."

The needs of NPDB patients and families are complex and multilayered. In light of participants' medical experiences and frustrations, it is clear that an integrated multidisciplinary approach is in the best interest of patients.

This was the first investigation of psychological aspects of patients and families with NPDB. The data suggest that patients experience psychosocial distress throughout childhood, adolescence, and adulthood. This distress seems to be related to social isolation and loss of control. As NPDB patients are confronted with challenging medical conditions on a daily basis, providing these individuals with adequate support and care in all areas of their physical and psychological being is essential. Unquestionably, increased knowledge about the relationship between chronic medical conditions of NPDB patients and mental health may serve to enhance the quality and quantity of services currently available.

An important strength of this study is the use of a multidimensional approach to assess psychosocial adjustment. Data were gathered from multiple respondents, including adolescents, parents, and adult patients. Each of these reports contributes to the complex picture of psychosocial adjustment in the context of chronic illness. Using both quantitative and qualitative approaches to understand psychosocial adjustment strengthens the study design. The use of qualitative methods in the form of case studies and interviews offers a rich view of the lives of patients and their families.

Some methodological issues should be noted. The sample used in this investigation consisted of all Caucasian individuals. Therefore, it is hard to predict whether the experience of this sample is similar to that of a different ethnic cohort of NPDB patients and families. A possible confound may be due to the retrospective reporting that was necessary to obtain data regarding development and diagnosis. The unreliability of retrospective memory may impact the assessment of patients' and families' memories of being diagnosed and growing up with NPDB. In addition, the participants in this study were drawn from a support group, and therefore may not reflect the full spectrum of the disease in terms of its medical manifestations or emotional, social, and developmental impact. On the other hand, given the rarity of the disorder, it is reasonable to consider that many, if not most, families would be affiliated with this support group.

This study investigated the current psychosocial adjustment of parents and patients with NPDB. An area that warrants further study is the psychosocial impact of NPDB on siblings. One mother alluded to the fact that she "worries" about her NPDB daughter, often to the exclusion of her other children. An investigation of siblings will further illuminate the pitfalls and challenges that families face while caring for a chronically ill child.

The results of this investigation further awareness of the psychosocial impact of living with a chronic illness. Although most of the participants showed relatively good adjustment, areas of psychosocial maladjustment were highlighted through the interview and

quantitative findings. This study calls for a collaborative, multidisciplinary effort in the treatment of these unique patients and their families.

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