Psychosocial Functioning in Youth with Glycogen Storage Disease Type I

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Objective To assess the quality of life and psychosocial functioning among pediatric patients with Glycogen Storage Disease (GSD) types Ia and Ib. Methods Thirty-one youth with GSD types Ia and Ib and 42 healthy controls participated. Quality of life ratings from the GSD types Ia and Ib sample were compared with a previously reported clinical comparison sample. Children completed measures of quality of life, loneliness, family functioning, and sibling relationship quality (e.g., warmth, conflict). Parents completed measures of parental distress, parenting stress, child adaptive behavior, and child emotional and behavioral functioning. Results Quality of life was generally lower in youth with GSD relative to healthy controls but similar to those with a chronic illness. Children with GSD were rated as having more internalizing symptoms, social problems, and lower independent functioning relative to healthy controls. Parents reported greater distress and parenting stress relative to healthy controls. Conclusions The presence of GSD types Ia and Ib are associated with reduced quality of life and independent functioning, and elevated levels of internalizing distress and parental stress relative to healthy controls. Relative to their children, parents generally reported that their child was more impaired, which suggests the need for multiple informants during assessment and active parental involvement during psychological treatment. These points should be kept in mind when assessing and treating youth with this disease and their families as psychological interventions that target areas of concern (e.g., adherence, coping with having a chronic disease) may be helpful for improving child and family outcomes.

Key words adjustment; children; glycogen storage disease; quality of life.

The glycogen storage diseases (GSD) are caused by inherited enzyme deficiencies that regulate the synthesis or degradation of glycogen. There are 12 types of GSD that are classified according to the type of enzymatic defect and primary organs that are affected (see Wolfsdorf & Weinstein, 2003 for a review of each type). Patients with GSD type I lack glucose-6-phosphatase activity, and this enzymatic deficiency impairs all endogenous glucose production, since it catalyzes the final step in both gluconeogenesis and glycogenolysis. GSD type Ia is caused by deficient catalytic activity of the glucose-6-phosphatase system whereas GSD type Ib is caused by a deficiency in the glucose-6-phosphate transporter enzyme (Wolfsdorf & Weinstein, 2003). GSD type Ia involves the liver, kidney, and intestines whereas GSD type Ib primarily affects the liver. In one study, median ages of symptom onset were 6 months and 4 months for GSD Ia and Ib, respectively (Rake et al., 2002a).

Patients with GSD types Ia and Ib present with fasting hypoglycemia, hepatomegaly, growth retardation, and metabolic disturbances (Wolfsdorf & Weinstein, 2003). Additionally, patients with type Ib present with additional clinical manifestations of intermittent severe neutropenia and neutrophil dysfunction that may lead to the development of severe infections and inflammatory
bowel disease (Visser et al., 2002). Suboptimal control is prospectively associated with many serious complications (e.g., gout, hepatic adenomas, osteoporosis, platelet dysfunction, pulmonary hypertension, and renal failure; Visser et al., 2000), and long-term neurologic injury has been reported even in well-controlled patients with GSD type I (Melis et al., 2004). Prior to the 1970s, the mortality rate for GSD was extremely high. Although life expectancy for GSD type I has improved substantially with dietary and pharmacological treatment, long-term prognosis remains unclear given the numerous and variable complications associated with the disease, as well as the relatively short time that effective treatment has been available. In a retrospective study, analyzing hospital records of 288 patients with GSD (median age at data collection for patients with type Ia = 10.4 years and type Ib = 7.1 years), findings have indicated that hepatic adenomas and progressive renal disease are likely the two major causes of mortality in GSD type I patients (Rake et al., 2002a).

Improved understanding of the pathophysiology present in the various types of GSD has allowed for the creation of treatments, which have greatly improved patient prognosis and enabled the majority of patients to live independently into adulthood. While clearly a positive step, these treatments require meticulous adherence and planning, and likely exert an impact on quality of life. For example, patients with GSD type I typically require glucose monitoring and cornstarch therapy (i.e., a nutritional intervention involving the intake of uncooked cornstarch to maintain plasma glucose concentrations) every 4–5 h around the clock, and these requirements disrupt activities of daily living and continuous sleep for both the child and his/her parent(s). Also, dietary guidelines restrict the intake of certain substances such as lactose, fructose, and sucrose, which can be particularly troublesome for children with GSD who are surrounded by foods containing these substances. In addition to dietary management, there are numerous pharmacological treatments used to prevent common complications of GSD type I. For example, there are medications to prevent gout and urate nephropathy (allopurinol), to treat lactacidemia (bicarbonate), to treat anemia (iron), and to prevent deterioration of renal function (blood pressure lowering drugs) (Rake et al., 2002b). Furthermore, proper monitoring of GSD type I symptoms and associated complications requires numerous and relatively frequent medical procedures, including ultrasounds, blood draws, bone densitometry, and colonoscopies (Rake et al., 2002b; Visser et al., 2002).

Thus, systematically coordinating these elements to maximize disease management can be complicated, time-consuming, and requires good organization. In particular, given that dietary treatments differ among different age groups and that guidelines recommend medical procedures at different time points (e.g., physical examination every 3 months, ultrasound of spleen every 12 months), being aware of adjustments to the regimen and keeping track of regimen requirements is a demanding process for family members. Given the complexity of the regimen, youth often have difficulty adhering to their specific medical regimens (Drotar, 2000), which can led to long-term complications as highlighted earlier.

Similarly, given the numerous stressors and required adherence tasks for youth with GSD (as well as their parents), it is not surprising that emotional adjustment and quality of life may be impacted. In youth with other medical conditions, particularly those that involve constant adherence to treatment recommendations, general quality of life and emotional functioning have been lowered relative to healthy peers (Hysing, Elgen, Gillberg, Lie, & Lundervold, 2007; Varni, Burwinkle, Seid, & Skarr, 2003b; Varni, Burwinkle, Rapoff, Kamps, & Olson, 2004; Williams, Wake, Hesketh, Maher, & Waters, 2005). Although very little is known about GSD, studies on the psychosocial effects of other pediatric chronic illnesses (e.g., cancer, asthma, cystic fibrosis) have suggested that children with a chronic illness must cope with a variety of disease-related stressors (Barlow & Ellard, 2006). For example, youngsters with a serious chronic illness often struggle with low self-esteem, problems with school functioning, and difficulty participating in extracurricular activities (Vitulano, 2003).

In addition to the impact of a chronic illness on psychological and emotional functioning, research indicates that having a chronic illness also disrupts social and family functioning. Studies of peer relations among children with chronic illness have revealed mixed findings. Some studies indicate that children with a chronic illness are prone to peer problems due to the social stigma associated with disease (e.g., Kashikar-Zuck et al., 2007; Sandstrom & Schanberg, 2004; Storch et al., 2007), although one study found that children with cancer had higher levels of social acceptance compared with their healthy peers (Noll et al., 1999). Peer-related difficulties may be related to decreased medical adherence, with one study indicating that diabetes-related bullying was related to poorer self-management and adherence to glucose testing and dietary tasks (Storch...
et al., 2006). This finding is relevant to youth with GSD, due to the importance of medical adherence for both GSD and diabetes. In addition, problems that arise secondary to chronic illness (i.e., change in physical appearance, restrictions placed on activities) may further affect peer relations (Spirito, De Lawyer, & Stark, 1991) and contribute to distress. This is particularly salient for youth with GSD, due to the impact of the disease on their ability to participate in daily activities. One study on family functioning revealed that mothers who indicated higher illness-related impairment in their children had higher rates of overall psychological distress, and this finding was particularly true for mothers with low perceived self-efficacy (Silver, Bauman, & Ireys, 1995). Marital relationships may also be affected by increased caretaking responsibilities as well as by anxiety and guilt about the cause of the child’s disease (Raina et al., 2005). Furthermore, a family’s social life can be restricted; family activities can be more difficult to organize, and financial resources may be more strained (Ray, 2005; Sleed, Eccleston, Beecham, Knapp, & Jordan, 2005). These family disruptions are common among GSD patients, given the time-intensive treatment regimens that require constant monitoring and considerable parent involvement.

Results from past research on the psychosocial impact of chronic illness pave the way to an investigation of psychosocial issues that affect children with GSD and their families. To date, virtually nothing has been published on the psychosocial issues related to GSD, despite the significant stressors related to this diagnosis. Talente et al. (1994) reported that of 37 adults with GSD type Ia, 25 had completed high school, 20 were completing or had completed postsecondary education, 11 were married or divorced, and six experienced clinical depression. No other reports of psychosocial functioning were available. Rake et al. (2002a) highlighted that the life-long intensive treatment regimens, as well as the serious medical complications associated with GSD, constitute a major burden for both patients and their parents. Findings also demonstrated that some GSD patients were suffering from depression and mental disability. Studies in children and adults with GSD type II documented significant interference in the ability to participate in daily activities (e.g., studying, work, sports) as well as impairment in the ability to perform self-care tasks, but did not investigate other psychosocial variables (e.g., internalizing symptoms, socialization) (Hagemans et al., 2007; Haley, Fraga-La, Aseltine, Ni, & Skrinar, 2003; Haley, Fraga-La, & Skrinar, 2003).

With this in mind, the present study aimed to assess several dimensions of psychosocial functioning among pediatric patients with GSD types Ia and Ib. The following specific questions were addressed:

1. Do parent and child ratings of quality of life differ among youth with GSD types Ia and Ib, healthy controls, and youth with varied clinical conditions?
2. Do youth with GSD types Ia and Ib present with greater rates of internalizing and externalizing problems and loneliness relative to healthy controls?
3. Is the adaptive functioning of youth with GSD types Ia and Ib different than that of healthy controls?
4. Are rates of parental stress and psychiatric symptoms different in parents of youth with GSD types Ia and Ib relative to healthy control parents?
5. Are varied domains of family functioning (e.g., family warmth, conflict, quality of sibling relationship) different in the families of youth with GSD types Ia and Ib relative to healthy control families?

Method

Participants

Participants were obtained from two sources: (a) children and adolescents diagnosed with GSD types Ia and Ib (n = 31; male = 16) seen for clinical management appointments in the Glycogen Storage Disease Program at University of Florida and their parent(s); (b) healthy controls recruited by written advertisements that were placed in facilities frequented by children and parents (e.g., schools, pediatrician’s offices, local grocery stores; n = 42, male = 21) and their parent(s). Healthy controls were screened at their assessment for the presence of any chronic illness that required ongoing medical care. Any youth who had a current or past chronic illness was excluded from this study. No gender, age, or ethnic differences existed between groups (p > .05). Youth participants’ ages ranged from 3 to 25 years (M = 11.11, SD = 4.18 years). Although this age range may seem quite broad and thus limit interpretations of findings, such a large age range was sampled given the low prevalence of GSD, which makes collecting a sizeable sample challenging, as well as the limited research investigating psychosocial factors in this condition. Two percent of the sample represented children aged 3 and 4 years, 58% of the sample represented children aged 6–12 years, 36% of the sample represented children aged 13 and older.
13–18 years, and 4% of the sample represented individuals aged 20–25 years. The sample was largely Caucasian (90%), followed by Hispanic (4%), African American (1%), and “Other” (5%). GSD type Ia/Ib diagnoses were confirmed by the final author by demonstration of abnormal glucose-6-phosphatase activity on a liver biopsy or may mutation analysis demonstrating a mutation in the glucose-6-phosphate or glucose-6-phosphate transporter genes.

To compare quality of life to a clinical comparison sample, PedsQL (Pediatric Quality of Life Inventory) scores for the current sample of children with GSD were examined relative to previously reported scores of youth with a chronic health condition (Varni et al., 2003b). This sample consisted of 831 children ages 2–16 years (and their caregivers) from California who had participated in a statewide survey of pediatric population health outcomes (Varni et al., 2003b). Parents and/or children who were English, Spanish, Vietnamese, Korean, or Cantonese speaking were included in the statewide survey. The chronic health conditions represented by these data included asthma (43.0%), diabetes (1.2%), attention-deficit hyperactivity disorder (9.9%), and depression (6.4%). Additionally, 27.4% of parents reported the presence of another chronic health condition that was not explicitly listed in the survey. We used this sample with both physical illnesses and mental health concerns rather than other possible samples involving a specific chronic illness as it is unclear if GSD mirrors any one specific chronic illness and thus, we wanted to provide a fair test of QoL in this sample. We have chosen to label the sample obtained from the Varni et al. (2003b) study as “clinical comparison group” to reflect that this sample includes children with both physical as well as mental health problems.

**Measures**

**Pediatric Quality of Life Inventory**

The Pediatric Quality of Life Inventory version 4.0 measures health-related quality of life in healthy children and those with acute and chronic medical conditions and consists of parallel child (PedsQL; Varni, Seid, & Rode, 1999) and parent-proxy (PedsQL parent proxy) measures of children’s QoL. Each of the 23 items is rated on a 5-point scale with higher scores corresponding to better QoL. Four factor analytically derived scales are embedded within the PedsQL: (a) physical functioning (eight items); (b) emotional functioning (five items); (c) social functioning (five items); and (d) school functioning (five items). These scales are combined to yield physical (equivalent to the physical functioning domain), psychosocial (sum of emotional, social, and school functioning domains), and total health scales (all four domains). Extensive psychometric data support the validity and reliability of the PedsQL and PedsQL parent proxy across multiple clinical presentations (Bastiaansen, Koot, Ferdinand, & Verhulst, 2004; Varni et al., 2003b). Cronbach’s alphas for the PedsQL and PedsQL parent-proxy total scores in the current sample were .86 and .93, respectively.

**Asher Loneliness Scale (ALS)**

The ALS (Asher & Wheeler, 1985) is a 16-item scale that assesses feelings of loneliness (e.g., “I have nobody to talk to at school”) and social adequacy (e.g., “It’s easy for me to make new friends at school;” reverse coded). The ALS was positively related to negative peer nominations and negatively associated with positive peer nominations and play ratings, supporting the convergent and divergent validity of this measure (Asher & Wheeler, 1985). For the current sample, Cronbach’s alpha for the ALS total score was .87.

**Child Behavior Checklist (CBCL)**

The CBCL (Achenbach, 1991) is a parent rating scale that assesses internalizing and externalizing behavior problems, and total behavior problems in children between the ages of 4 and 18 years. Parents of children ages 4–18 years completed the CBCL. Extensive reliability and validity data have been reported on the CBCL (Achenbach, 1991). Cronbach’s alpha for the CBCL total score in the current sample was .95.

**Brief Symptom Inventory (BSI)**

This is a 53-item measure that provides an assessment of distress in adults 18+ years on nine subscales, as well as three global scales including Global Severity Index (GSI), Positive Symptom Distress, and Positive Symptom Total. The GSI summarizes the overall level of psychological distress and will be used in the present study. Individuals are asked to respond to each item in terms of “how they have been feeling during the past 7 days.” In this study, only parents completed the BSI (Derogatis & Melisaratos, 1983). Cronbach’s alpha for the GSI score was .96.

**Pediatric Inventory for Parents (PIP)**

The PIP (Streisand, Braniecki, Tercyak, & Kazak, 2001) is a 42-item parent self-report rating of stress associated with caring for a child with a medical illness. Items on the PIP are grouped into one of four domain scales (Communication, Medical Care, Role Functioning, and Emotional Functioning) and rated as to both the item’s frequency [Pediatric Inventory for Parents-frequency (PIP-F)] over the last week and the level of difficulty [Pediatric Inventory for Parents-frequency (PIP-F)] over the last week and the level of difficulty...
Parents-difficulty (PIP-D)] associated with it. Excellent psychometric properties have been reported in samples of youth with cancer (Streisand et al., 2001; Streisand, Kazak, & Tercyak, 2003), type 1 diabetes (Lewin et al., 2005), and obesity (Ohleyer et al., 2007). Cronbach’s alpha for the PIP-F and PIP-D total scores were .98 and .97, respectively.

McMaster Family Assessment Device (FAD)
The FAD (Epstein, Baldwin, & Bishop, 1983) is a 60-item self-report measure of family functioning that consists of six scales (Problem Solving, Communication, Roles, Affective Responsiveness, Affective Involvement, and Behavior Control) and an overall General Functioning scale. The FAD has been used as a measure of family functioning in a variety of pediatric samples including children with disabilities and medical illness. Items are generally geared to respondents who are ~10 years and older. In the present study, participants ages 7 and above completed this measure only if it was clear that they were able to understand questions (as assessed through their ability to read and accurately respond to other measures).

The FAD differs from the PIP in that the latter is a measure of stress related to caring for a child with a chronic illness, whereas the former is an index of family functioning across a variety of domains. Cronbach’s alpha for the FAD parent-report and child-report General Functioning scales were .89 and .91, respectively.

AAMR Adaptive Behavior Scale-School:
Second Edition (ABS-S2)
The ABS-S2 (Lambert, Nihira, & Leland, 1993) is an adaptive behavior scale designed to measure children’s personal and community independence and skills and adjustment. The ABS-S2 assesses both behavioral and affective competencies across five factors: Personal Self-sufficiency, Community Self-sufficiency, Personal-social Responsibility, Social Adjustment, and Personal Adjustment. This study examined subscales of the ABS-S2 that assess the child’s degree of personal independence and personal responsibility in daily living. Specific subscales used in this study were: Independent Functioning, Physical Development, Economic, Language, Numbers and Time, Prevocational/vocational, Self-direction, Responsibility, and Socialization. Internal consistency was not assessed for the ABS-S2 given that this statistic would not be appropriate given the response format.

Sibling Relationship Questionnaire (SRQ)
The SRQ (Furman & Buhrmester, 1985) is a 39-item self-report measure that assesses children’s perceptions (child-report version) of their relationship with their closest-in-age sibling, including factors related to perceived warmth, relative status/power, conflicts, and rivalry. The SRQ has been used in youth as young as 7 years (Labay & Walco, 2004). In the present study, only participants ages 7 years and above completed this measure. The parent-report SRQ assesses the parent’s perception of the target child’s relationship with their closest-in-age sibling on the same factors. Cronbach’s alpha for the parent-report of the warmth, conflicts, and rivalry factors were .95, .85, and .73, respectively. Cronbach’s alpha for the child-report of the warmth, conflicts, and rivalry factors in the current sample were .95, .88, and .37, respectively. Given the low alpha, the child-rated rivalry factor was not included in subsequent analyses.

Participants
The University of Florida institutional review board and General Clinical Research Center granted permission to conduct this research, and written parental consent and child assent (for youth younger than 18 years) were obtained for each of the participants prior to the administration of study measures. Those participants with GSD type Ia/Ib who were 18 years or older provided written consent; their parent provided written consent for their own participation. Parents and children were told that their involvement was voluntary, and they could refuse permission without negative consequences of any kind. Instructions were provided for each measure by a trained research assistant, who was available to answer questions. Each family was offered $50 compensation for their participation.

Results
Comparison of Quality of Life in Youth with GSD, a Clinical Comparison Group, and Healthy Controls

Given that there were no published data from which to estimate effect size related to mean differences using a sample of youth with GSD, we calculated power based on an effect size estimated from the difference between the means of the parent-proxy report of the PedsQL in a sample of youth with type 1 and type 2 diabetes and a sample of healthy children reported in the Varni et al. (2003a) study. After calculating a Cohen’s d for effect size, a sigma estimate was made using a weighted standard deviation (SD = 12.89). G*Power provided a d-estimate of effect size of .86. At α = .05 and power = .80, G*Power estimated that a minimum sample of 36 (two groups of 18) would be needed to detect this effect size.
QoL scores for the GSD sample, the clinical comparison group sample, and the healthy control sample are reported in Table I. Given the relatively small sample size and the preliminary nature of these data, we did not correct for the increased possibility of Type I error. Therefore, the alpha level for significance was set at $p < .05$.

Independent-sample t-tests were used to compare scores on the PedsQL for the GSD sample and the healthy control sample (see Table II). Differences in the number of youth completing certain measures were a function of young age, such that children younger than the age of 5 years did not complete the PedsQL. Findings revealed that QoL scores for the GSD sample were significantly lower (indicating worse QoL) than those for healthy controls on the PedsQL total score and the domains of physical health, psychosocial health, and social functioning. Results revealed no significant differences between QoL scores for the GSD sample and the healthy control sample on the PedsQL domains of emotional functioning and school functioning. QoL scores for the GSD sample were significantly lower than those of healthy controls for the PedsQL parent-proxy total score and the domains of physical health, psychosocial health, emotional functioning, social functioning, and school functioning.

One-sample t-tests were used to compare the GSD type I sample with the clinical comparison group sample (Varni et al., 2003b). Results indicated that the two groups did not differ significantly on either child-report or parent proxy-report of QoL.

### Comparison of Behavior Problems and Adaptive Functioning in Youth with GSD and Healthy Controls

Table III presents scores obtained from the CBCL, ALS, and ABS-S2 for the GSD sample and the healthy control sample. Using independent-sample t-tests, significant group differences were found on the CBCL total score, the CBCL Internalizing scale, and the CBCL Social

### Table I. Descriptive Statistics and Bivariate Analyses of Quality of Life

<table>
<thead>
<tr>
<th></th>
<th>Healthy control sample</th>
<th>Clinical comparison group sample</th>
<th>GSD sample</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>(n = 42)</td>
<td>(n = 831)</td>
<td>(n = 31)</td>
</tr>
<tr>
<td>PedsQL</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total score</td>
<td>80.90</td>
<td>74.16</td>
<td>71.65</td>
</tr>
<tr>
<td>M</td>
<td>SD</td>
<td>M</td>
<td>SD</td>
</tr>
<tr>
<td>Physical functioning</td>
<td>88.68</td>
<td>79.47</td>
<td>75.63</td>
</tr>
<tr>
<td>Psychosocial health</td>
<td>77.38</td>
<td>71.32</td>
<td>70.33</td>
</tr>
<tr>
<td>Emotional functioning</td>
<td>71.00</td>
<td>69.32</td>
<td>68.20</td>
</tr>
<tr>
<td>Social functioning</td>
<td>86.33</td>
<td>76.36</td>
<td>71.40</td>
</tr>
<tr>
<td>School functioning</td>
<td>75.34</td>
<td>68.27</td>
<td>71.40</td>
</tr>
<tr>
<td>PedsQL parent proxy</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total score</td>
<td>92.36</td>
<td>73.14</td>
<td>74.88</td>
</tr>
<tr>
<td>Physical functioning</td>
<td>95.27</td>
<td>76.99</td>
<td>76.45</td>
</tr>
<tr>
<td>Psychosocial health</td>
<td>91.38</td>
<td>71.04</td>
<td>74.36</td>
</tr>
<tr>
<td>Emotional functioning</td>
<td>88.05</td>
<td>71.08</td>
<td>76.93</td>
</tr>
<tr>
<td>Social functioning</td>
<td>95.61</td>
<td>75.06</td>
<td>74.23</td>
</tr>
<tr>
<td>School functioning</td>
<td>90.49</td>
<td>65.58</td>
<td>71.92</td>
</tr>
</tbody>
</table>

$>$"$" signifies that the sample’s scores were significantly ($p < .05$) greater than another sample’s scores; "$=$" signifies that the sample’s scores were not significantly different from another sample’s scores.
Problems subscale, such that scores for the GSD sample were significantly higher (indicating more problems) than those for the healthy control sample. No significant differences were found between groups on the ALS. Additionally, scores for the GSD sample were significantly lower (indicating worse functioning) than those for the healthy controls on the ABS-S2 Independent Functioning subscale and the Economic subscale. The latter subscale assesses tasks like managing money, budgeting, and shopping, whereas the former assesses levels of prevocational/vocational activity (i.e., school- or job-related skills). All other differences were nonsignificant.

**Comparison of Parental and Family Functioning in Youth with GSD and Healthy Controls**

Table IV presents scores obtained from parent ratings on the BSI Global Stress Index, PIP, FAD, and SRQ, and child ratings on the FAD and SRQ for the GSD sample and the healthy control sample. Using independent-sample t-tests, significant group differences were found for ratings of parental stress associated with caring for a child with a medical illness, as well as general parental distress (e.g., anxiety, depressive symptoms). Specifically, scores for the parents of the GSD sample were significantly higher (indicating more stress and distress) than those for the parents of the healthy control sample on the BSI-GSI. All scores for the SRQ were nonsignificant (p’s > .05; See Table IV). Additionally, results revealed no significant differences between groups on both parent-report and child-report scores on the FAD (p’s > .05; See Table IV).

**Discussion**

The present study examined psychosocial functioning and parental distress in youth with GSD types Ia and Ib. Similar to previous research on children with chronic health conditions (Hysing et al., 2007; Varni et al., 2003b, 2004; Williams et al., 2005), results generally indicated that youth with GSD experience lower quality of life in several areas when compared with healthy children. Youth with GSD reported similar quality of life issues as children with other chronic health and mental health conditions (i.e., Varni et al., 2003b). There were no differences between these groups with regard to child or parent-rated quality of life. Of note, families of youth with GSD reported general family functioning (i.e., functioning unrelated to the child’s condition, such as communication, problem solving, etc.) equivalent to that reported by families of healthy youth. This suggests that, despite the significant stress related to maintaining a complicated medical regimen, restrictions on activities, and disruption due to physician visits, these families are able to maintain adequate communication, problem solving, role distinction, affection, and control. It would be interesting for future efforts to examine the factors that help buffer against distress in families with a child with GSD. However, like other families of children with chronic illness (Silver et al., 1995), parents of youth with GSD did report significantly increased frequency and severity of distress related to caring for a child with special needs, suggesting a potential area of needed intervention with these families.

### Table III. Descriptive Statistics and Bivariate Analyses of Behavior Problems and Adaptive Functioning

<table>
<thead>
<tr>
<th></th>
<th>Healthy control sample (n = 42)</th>
<th>GSD sample (n = 31)</th>
<th>t-test</th>
<th>Effect size (CI)</th>
<th>CI for means</th>
</tr>
</thead>
<tbody>
<tr>
<td>CBCL</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total score</td>
<td>10.95</td>
<td>19.65</td>
<td>2.30</td>
<td>.03</td>
<td>.58 (.11 to 1.06)</td>
</tr>
<tr>
<td>Internalizing</td>
<td>2.81</td>
<td>5.92</td>
<td>2.39</td>
<td>.02</td>
<td>.60 (.13 to 1.08)</td>
</tr>
<tr>
<td>Externalizing</td>
<td>3.08</td>
<td>4.89</td>
<td>1.48</td>
<td>.14</td>
<td>.38 (−.09 to .84)</td>
</tr>
<tr>
<td>Social problems</td>
<td>.68</td>
<td>2.46</td>
<td>4.28</td>
<td>&lt;.01</td>
<td>1.08 (.58 to 1.57)</td>
</tr>
<tr>
<td>ALS total score</td>
<td>54.45</td>
<td>53.17</td>
<td>−1.10</td>
<td>.28</td>
<td>−.29 (−.76 to .17)</td>
</tr>
<tr>
<td>ABS-S2 independent functioning</td>
<td>103.59</td>
<td>97.42</td>
<td>2.19</td>
<td>.03</td>
<td>.56 (.09 to 1.04)</td>
</tr>
<tr>
<td>Physical development</td>
<td>23.71</td>
<td>23.60</td>
<td>.34</td>
<td>.73</td>
<td>.09 (−.38 to .55)</td>
</tr>
<tr>
<td>Economic</td>
<td>14.63</td>
<td>9.83</td>
<td>2.40</td>
<td>.02</td>
<td>.62 (.15 to 1.10)</td>
</tr>
<tr>
<td>Language</td>
<td>40.49</td>
<td>40.32</td>
<td>.19</td>
<td>.85</td>
<td>.05 (−.42 to .51)</td>
</tr>
<tr>
<td>Numbers and time</td>
<td>12.98</td>
<td>12.96</td>
<td>.03</td>
<td>.97</td>
<td>.01 (−.45 to .47)</td>
</tr>
<tr>
<td>Provocational/vocational</td>
<td>9.95</td>
<td>9.04</td>
<td>2.00</td>
<td>.05</td>
<td>.49 (−.02 to .96)</td>
</tr>
<tr>
<td>Self-direction</td>
<td>21.59</td>
<td>20.72</td>
<td>1.13</td>
<td>.26</td>
<td>.28 (−.19 to .75)</td>
</tr>
<tr>
<td>Responsibility</td>
<td>8.56</td>
<td>8.17</td>
<td>1.58</td>
<td>.12</td>
<td>.40 (−.07 to .87)</td>
</tr>
<tr>
<td>Socialization</td>
<td>25.08</td>
<td>24.80</td>
<td>.52</td>
<td>.61</td>
<td>.14 (−.33 to .60)</td>
</tr>
</tbody>
</table>
Interestingly, youth with GSD acknowledged greater difficulties than healthy children in overall quality of life, physical functioning, and social functioning, which suggests that the impact of disease-related stressors (Barlow & Ellard, 2004) may be most prominent in these areas for this population. Certainly, as suggested in previous research (Spirito et al., 1991), the restrictions on physical activity for these patients would influence their perceptions regarding their physical functioning. It is likely that activity and food restrictions, as well as the complicated cornstarch treatment regimen, also interfere with social functioning by highlighting the differences among youth with GSD and healthy youth in observable ways. It is noteworthy that youth with GSD type I denied having greater difficulties than healthy children in psychosocial health, emotional functioning, or school functioning, suggesting that they have developed adaptive ways of managing stressors that might impact their functioning in these areas. For example, with regard to school functioning, students who are unable to participate in physical activity (e.g., gym class, recess) may spend this time completing homework or other school-related tasks.

On the other hand, like parents of children with other chronic health and mental health conditions (Bastiaansen et al., 2004; Varni et al., 2003b; Williams et al., 2005), the parents of youth with GSD type I reported that their children had greater difficulties in all of these areas. Studies examining the discrepancy between parent and child reports of child well-being have been well-documented in the literature on chronic pediatric conditions (Levi & Drotar, 1999; Klassen, Miller, & Fine, 2006; Robitail et al., 2007). The discrepancies suggest three possibilities: (a) youth with a chronic illness do not experience as much social/emotional distress or difficulty at school as their parents think (i.e., parents are overly sensitive to their children’s level of difficulty), (b) youth with chronic illness are hesitant to report difficulties in these areas, or (c) youth with chronic illness do not have good insight into the
severity of their impairments. Some attempts have been made to clarify this discrepancy (Cremeens, Eiser, & Blades, 2006), but more work is needed in this area.

This same pattern of reporting (i.e., parents endorsing elevated child adjustment concerns, children reporting few concerns) was displayed with other measures. For example, parents of youth with GSD reported greater levels of internalizing problems and social problems (on the CBCL) than did parents of healthy youth. But, youth with GSD did not report greater loneliness than their healthy peers. Although these measures do not assess exactly the same construct, child-reported loneliness would be expected to be related to internalizing symptoms and social problems and can serve as an adequate proxy. As a result, this may reflect differences in perception (e.g., the child has 1–2 close friends or siblings and thus does not feel lonely; whereas the parent sees that the child has only 1–2 friends and worries that the child is not functioning on an optimal social level). Alternatively, it is possible that youth with GSD have poor insight into their social/emotional difficulties and may not be aware of this impairment, and/or responded to measures in a socially desirable manner. Finally, it may reflect a generalization of the parents’ overall distress. Like parents of other children with chronic illness (Silver et al., 1995), parents of youth with GSD reported more psychosomatic symptoms than did parents of healthy children, and also reported greater frequency and severity of parental stress. Perhaps these parents are more vigilant and attentive to their children’s needs, challenges, and difficulties, and exert more effort in attempting to manage these issues.

Finally, parents of youth with GSD also reported that their children were less able to function independently than healthy children. Given the complicated medical regimen that these children must maintain and the difficulty that many children have managing a medical regimen independently (Drotar, 2000), these results were not surprising. However, the parents of youth with GSD reported that their children were less able to manage money, budget, and shop independently and showed lower levels of prevocational/vocational activity (i.e., school- or job-related skills). It is unclear whether these differences result from decreased ability or decreased opportunity to engage in these activities. Perhaps parents of youth with GSD are more restrictive regarding their children’s activities, which may be due to concerns about their child’s health or self-management of the treatment regimen. Future studies should examine this question.

Some limitations of the present study should be noted. First, the sample was relatively small, which limited our ability to analyze the effects of demographic (e.g., age, gender) and illness-related variables (e.g., disease duration) on the constructs measured in this study. However, given that the estimated prevalence of GSD (Types I–IV) is only about 0.00004% (Wolfsdorf & Weinstein, 2003), this study actually encompasses a relatively large percentage of the population of individuals with GSD type I. Second, the healthy control sample of children was relatively homogeneous with regard to demographic characteristics; though it did not differ significantly from the make-up of the GSD sample. Third, the assessments in this study consisted of solely self-report (or parent-proxy-report) measures. Thus, it is possible that the results were influenced by differences in response patterns. Future studies should attempt to incorporate other methods of assessment (e.g., teacher report, clinician report, and objective data) in order to obtain a more balanced view.

**Clinical Implications**

Within these limitations, this study provides preliminary data regarding social, emotional, behavioral, and familial functioning in families of youth with GSD types Ia and Ib. Given that parents of youth with GSD type I reported significant distress related to caring for their child with a chronic illness, it is likely that these families would benefit from consultation with a pediatric psychologist in order to learn adaptive ways to manage this extra responsibility and monitor psychosocial functioning. In particular, parents may benefit from learning strategies to manage their own personal distress while attempting to balance the burden of helping their child and maintaining a “normal” lifestyle. Children with GSD type I would likely benefit from learning ways to modify their participation in certain activities (e.g., physical games) in order to remain included while adhering to their regimen and restrictions. In addition, it is possible that parents and their children with GSD type I could benefit from professional assistance in determining appropriate opportunities for the child to participate in activities independently. This would help to ensure that the parents are not being overly protective and that the youth can grow in developmentally appropriate ways.

**Conflicts of interest**: None declared.

Received September 21, 2007; revisions received February 5, 2008; accepted February 11, 2008
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