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# Psychosocial Functioning in Youth with Barth Syndrome

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## Abstract

This pilot study assessed the quality of life and psychosocial functioning of pediatric patients with Barth Syndrome. Thirty-four boys with Barth Syndrome and 22 healthy male controls were administered a measure of verbal ability and completed measures of quality of life, loneliness, perceived peer support, and sibling relationship quality. Parents completed measures of parental distress, parenting stress, child academic functioning, child adaptive behavior, and child emotional and behavioral functioning. Quality of life ratings were consistently lower in youth with Barth Syndrome relative to both healthy controls and a previously reported sample of youth with cardiac disease. Compared to healthy controls, children with Barth Syndrome were rated as having more internalizing and externalizing symptoms, social problems, loneliness, and lower independent functioning. Parents of boys with Barth Syndrome reported greater distress and parenting stress relative to healthy controls. In addition, parents reported a significant need for academic accommodations, given their son's illness and associated impairments. Boys with Barth Syndrome and their parents appear to be affected by the presence of the illness in numerous ways. Results suggest the need for interventions aimed at helping children and families cope with illness-related stressors to enhance quality of life and overall functioning.

### Keywords

Barth Syndrome; Children; Chronic Illness; Quality of life; Psychosocial functioning

Barth Syndrome is an X-linked disorder that occurs in approximately 1 in 300,000 births (Kelley, 2007) with the principle features of cardioskeletal myopathy, neutropenia, and growth retardation (Barth et al., 2004). Cardiomyopathy usually presents within the first two years of life, with most boys having left ventricular dysfunction and presenting with symptoms of heart failure. Neutropenia is often cyclic and predisposes the boys to serious bacterial infections. Patients with Barth Syndrome usually manifest skeletal muscle weakness and are often delayed

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in gross motor development (Ades et al., 1993; Barth, Wander, & Vreken, 1999). Finally, Barth Syndrome is characterized by marked deceleration in growth, beginning in the first year of life (Ino et al., 1988; Kelley et al., 1991). Indeed, patients with Barth Syndrome are considerably below average in height and weight, with most patients falling at least two standard deviations below normal stature by two years of age (Barth et al., 2004). Although survival rates for patients with Barth Syndrome have increased considerably since the identification of the clinical phenotype, the mortality rate remains high relative to other pediatric illnesses (Barth et al., 1983; Barth et al., 2004). Alertness to the diagnosis and early treatment of infections and cardiomyopathy have been credited for increased survival rates.

Identification of the clinical phenotype of Barth Syndrome has also generated research investigating the genetic and biological aspects of the illness. However, only two reports (Mazzocco & Kelley, 2001; Mazzocco, Henry, & Kelly, 2007) have examined psychosocial issues in boys with Barth Syndrome, despite considerable clinical evidence that the boys and their families endure a number of significant psychosocial challenges. The Mazzocco and Kelley (2001) study examined profiles on the Child Behavior Checklist (Achenbach, 1991) and Conners' Parent Form (Conners, 1997) among five boys with Barth Syndrome. Relative to a non-clinical sample of boys, social concerns were reported by parents of boys with Barth Syndrome significantly more frequently. In addition, relative to a non-clinical and age/gender/grade matched sample, significantly more academic and cognitive concerns were noted in the Barth Syndrome sample, a finding confirmed by a more recent study by the same group (Mazzocco et al., 2007). It is noteworthy, however, that the Mazzocco and Kelly (2001) study was relatively small in scope and that score dispersion was significant, with some boys showing few problem areas on psychosocial measures, and others endorsing multiple concerns. Similarly, the Mazzocco et al. (2007) study only examined cognitive and academic variables limiting inferences related to psychosocial functioning.

Assessing the psychosocial effects of Barth Syndrome on the affected child and his entire family is critically important given the serious implications of the illness. Barth Syndrome has the potential to negatively affect many important domains of functioning. For example, physiological aspects of the illness itself, medications and other forms of treatment, frequent hospitalizations, disruptions in daily activities, and alterations in family relationships are some of the potential factors that may negatively affect the psychosocial adjustment of boys with Barth Syndrome and their families. Anecdotal accounts from clinicians and parents of boys with Barth Syndrome have suggested that these families face an array of illness-related stressors that may weigh heavily on their ability to cope effectively. For example, many boys experience socialization difficulties and bullying, which may be attributed to features of their illness (e.g., small stature, inability to participate in certain play activities that require physical exertion). Some boys also experience school problems, ranging from poor attendance to mild visual-spatial and motor learning impairments that impede learning (Mazzocco & Kelley, 2001; Mazzocco et al., 2007).

In addition to the effects that the illness has on the child, parents of boys with Barth Syndrome have also reported significant distress related to their child's illness. This finding is consistent with other pediatric chronic illnesses (Plant & Sanders, 2007). Not surprisingly, the added financial, psychological, and social stressors of raising a child with a chronic illness can weigh heavily on parents' ability to effectively cope (Montagnino & Maruicio, 2004). Many parents with children who have chronic illnesses endorse feelings of distress linked to their child's condition (Raina et al., 2005).

Siblings of boys with Barth Syndrome may also be impacted by their brother's illness. Because boys with Barth Syndrome require a substantial amount of parental attention, siblings may feel isolated from their family and resentful of their siblings' illness, a finding that has been shown

in other research (Barlow & Ellard, 2006). In addition, because the risk of death due to complications from the illness is relatively high, the boys and their family members may experience sadness and anxiety surrounding their child/sibling's health status and may have difficulty contending with demands resulting from the illness. Female siblings may feel distress knowing that they might be carriers of the illness. Given the negative psychosocial consequences associated with having Barth Syndrome for affected boys and their families, research was needed to identify areas in which psychosocial interventions may be effective in decreasing distress and improving quality of life of families and children with Barth Syndrome.

As noted, little attention has been given to the psychosocial aspects of Barth Syndrome in affected boys or their family members. Given this, we examined several specific areas of psychosocial adjustment:

1. Are parent- and child-rated quality of life for a sample of boys with Barth Syndrome different than for a non-clinical control sample, and a sample of youth with a cardiac disease (Varni et al., 2007)? A comparison sample of youth with cardiac disease was used given that cardiac issues are a significant part of the Barth Syndrome phenotype. Thus, we sought to examine the extent to which quality of life in boys with Barth Syndrome was impacted over and above having cardiac problems in isolation.
2. Are internalizing and externalizing symptoms and loneliness more prevalent in boys with Barth Syndrome than in healthy male controls?
3. Is the adaptive functioning of youth with Barth Syndrome different than that of healthy male controls?
4. Are rates of parental stress and psychiatric symptoms different in parents of youth with Barth Syndrome than in parents of healthy boys?
5. Is the quality of the sibling relationship in families of boys with Barth Syndrome different from sibling relationship among healthy male controls?
6. What types of academic stressors do youth with Barth Syndrome face?

## Method

### Participants

Participants consisted of families of boys with Barth Syndrome ($n = 34$) and families of healthy controls who were recruited via written advertisements ($n = 22$). The participants with Barth Syndrome were seen at the biannual conference of the Barth Syndrome Foundation held in Florida. Prior to this conference, families were offered the opportunity to participate in a number of research studies that were approved by the Foundation. If they expressed interest in the study, they scheduled a meeting with the investigators to learn more and review the relevant consent procedures. In the instance that the family remained interested, written consent and assent were obtained (if the child was old enough to provide assent) and the study commenced. If the boy with Barth Syndrome was over the age of 18 years ($n = 8$), he provided consent for his own participation and his parent provided consent for his or her study involvement. Healthy controls were assessed at the onset of their study visit for the presence of any chronic illness that required ongoing medical care. Participants with a past or current chronic illness were excluded. No age or ethnic differences existed between groups ($p > .05$). A broad age range for participants ($M = 11.21$, $SD = 6.37$ years, range = 2–25) was used, given the low prevalence rates of Barth Syndrome (i.e., there are under 100 cases known in the world), the preliminary nature of this work (i.e., there is very little available psychosocial information), and to inform future research. Given this large age range, certain measures were not applicable to some of the participants, and therefore, there is variability in the number of participants who

completed each measure. Furthermore, some of the measures of family functioning were not completed by all participants due to either (1) failure of the participant to complete all measures in the study packet or (2) the absence of a sibling in the family composition. The majority of the sample was Caucasian (91%), followed by Hispanic (4%), African-American (2%), and 'Other' (3%).

## Measures

**Pediatric Quality of Life Inventory (PedsQL; Varni et al., 1999)—**The Pediatric Quality of Life Inventory Version 4.0 measures health-related quality of life (HRQOL) in healthy children and those with acute and chronic medical conditions. The child (PedsQL) and parent (PedsQL Parent Proxy) versions both measure the child's HRQOL over the last month, with separate versions for children 8–12 years of age and teens 13–18 years of age, as well as respective proxy versions for parents of each age group. This 23-item module assesses four factorially derived scales (Physical – 8 items, Emotional – 5 items, Social – 5 items, and School – 5 items) which generate three summary scores: (a) Total Scale Score – 23 items, (b) Physical Health Summary Score – 8 items, and (c) Psychosocial Health Summary Score – 15 items. All items are statements rated on a 5-item Likert scale ranging from "Never" to "Almost Always," with increasing numbers indicating more positive quality of life. In the current study, Cronbach's α values for score reliability for the PedsQL Total Scale Score (α = .94) and the Parent-Proxy PedsQL Total Scale Score (α = .98) were within acceptable limits for group comparisons. Extensive psychometric data support the validity and score reliability of the PedsQL and PedsQL Parent Proxy across multiple clinical presentations (e.g., Bastiaansen et al., 2004; Varni et al., 2003).

**Asher Loneliness Scale (ALS; Asher & Wheeler, 1985)—**This 24-item questionnaire is comprised of statements to which the child rates their agreement on a 5-point scale ranging from "That's not true at all about me" to "That's always true about me." Of the 24 items, which include 8 filler items focusing on hobbies and preferred activities, 16 have been factor analyzed to be loaded on the "loneliness and social dissatisfaction" factor. Those items focus on: (1) children's feelings of loneliness, (2) feelings of social adequacy versus inadequacy, and (3) subjective estimations of peer status. The collective 16 items have been found to have excellent internal consistency score reliability (Cronbach's α = .90; Asher et al., 1985). The Cronbach's alpha for the current sample was .90. In other research, the ALS was positively related to negative peer nominations and negatively associated with positive peer nominations and play ratings, providing support for the convergent and divergent validity of this measure (Asher & Wheeler, 1985).

**Perceptions of Peer Support Scale (PPSS; Kochenderfer & Ladd, 1997)—**The PPSS is a 4-item self-report measure that assesses a child's perception of peer victimization. The measure asks the child to rate the extent to which he has experienced four types of peer aggression: verbal, physical, general and indirect. Cronbach's alpha for the PPSS Total Score in the current study was .81.

**Brief Symptom Inventory (BSI; Derogatis & Melisaratos, 1983)—**This is a 53-item measure that provides an assessment of distress in adults above 18 years of age on 9 subscales, as well as 3 global scales including Global Distress (GSI), Positive Symptom Distress, and Positive Symptom Total. Items are rated on a 5-point Likert-type scale ranging from 1 ("Not At All") to 5 ("Extremely"). Individuals are asked to respond to each item in terms of "how they have been feeling during the past 7 days." The score reliability of the BSI is acceptable, with coefficients ranging from .68 to .91 (Peterson, 1989). In the current study, Cronbach's alpha for the Global Distress Index was .94.

**Pediatric Inventory for Parents (PIP; Streisand, Braniecki, Tercyak, & Kazak, 2001)—**The PIP 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 on the frequency and difficulty of the stressor over the last week. Internal consistency score reliability in the current study was excellent, with both the PIP-Difficulty (PIP-D) Score and the PIP-Frequency (PIP-F) Scores displaying Cronbach's alphas of .96. Additionally, the PIP has demonstrated good construct validity, being significantly correlated to measures of both state anxiety and parenting stress (Streisand et al., 2001).

**Sibling Relationship Questionnaire (SRQ; Furman & Buhrmester, 1985)—**The SRQ is a 39-item self-report measure that assesses children's perceptions of their relationship with their closest-in-age sibling. It assesses perceived Warmth, Relative Status/Power, Conflict, and Rivalry. In the current study, the Warmth, Conflict, and Rivalry subscales were used. Individuals were asked to rate features of relationships to their own sibling relationship on a 5-point Likert-type scale ranging from "Hardly at All" to "Extremely Much." In the current study, Cronbach's alpha for the SRQ Warmth, Conflict, and Rivalry scales were .95, .93, and .79, respectively. Psychometric testing of the SRQ has shown Cronbach's coefficients ranging from .71 to .81 (Furman & Buhrmester, 1985). Additional psychometric evaluation by Furman and Buhrmester (1985) also found that the SRQ showed acceptable test-retest reliability, ranging from .58 to .86.

**Child Behavior Checklist (CBCL; Achenbach, 1991)—**Depending on child age, parents completed the CBCL/1½ to 5 years or the CBCL/6–18 years. Parents of youth over 18 years completed the CBCL/6–18 years. Scores were standardized across forms. The CBCL is a parent rating scale that assesses internalizing and externalizing behavior problems, and total behavior problems in children between the ages of 1½ and 18 years. Extensive reliability and validity data have been reported on the CBCL (Achenbach & Rescorla, 2001). In the current study, Cronbach's alpha for the CBCL Total Score was .94.

**AAMR Adaptive Behavior Scale-School: Second Edition (ABS-S2; Lambert, Nihira, & Leland, 1993)—**The ABS-S2 is an adaptive behavior scale that measures 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 assessed 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.

**Peabody Picture Vocabulary Test – Third Edition (PPVT-III; Dunn & Dunn, 1997)—**The PPVT-III is an individually administered, norm-referenced test designed to assess receptive vocabulary and verbal ability in individuals ages 2 ½ to 90 years. It is a non-verbal test, only requiring the ability to point to one of four pictures matching a given target word. The PPVT-III was used to insure an adequate level of verbal receptiveness in participating subjects (defined as a scaled score ≥ 80) and to provide a broad measure of intellectual functioning. This measure also was chosen because of the ability to assess all participants, regardless of age. Alpha reliability scores range from .86 to .98 for the two parallel forms of this measure (Dunn & Dunn, 1997). Further, research by Dunn and Dunn (1997) demonstrated that scores on the PPVT-III are correlated to verbal intelligence quotient (VIQ) scores on the

Wechsler Intelligence Scale for Children – Third Edition (WISC-III; Wechsler, 1991) with scores above .90 for both parallel forms.

### Procedures

All study procedures were approved by the University of Florida institutional review board. After explaining the purpose of the study, written parental consent and child assent (for youth younger than 18 years) were obtained from study participants. Participants were instructed that involvement was voluntary and confidential, there were no consequences for refusing to participate, and participants would receive $40 compensation for their time. Thereafter, instructions for measures were provided by a trained research assistant and participants completed relevant measures.

## Results

### Comparison of Quality of Life in Youth with Barth Syndrome, a Cardiac Disease Comparison Group, and Healthy Male Controls

Quality of Life scores for the Barth Syndrome sample, the cardiac disease sample (Varni et al., 2007), and the healthy control sample are presented in Table 1. Due to 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 $p < .05$.

We conducted independent-sample *t*-tests to compare scores on the PedsQL for the Barth Syndrome sample and the healthy male control sample (see Table 2). Results indicated that quality of life scores for the Barth Syndrome sample were significantly lower (indicating worse quality of life) than those for healthy controls on the PedsQL total score, as well as the domains of physical health, psychosocial health, emotional functioning, social functioning, and school functioning. Regarding the PedsQL Parent-Proxy scores, quality of life scores for the Barth Syndrome sample were significantly lower than those of healthy controls for the total score and the domains of physical health, psychosocial health, emotional functioning, social functioning, and school functioning.

We conducted one-sample *t*-tests to compare the Barth Syndrome sample with males from the cardiac disease sample (Varni et al., 2007). Findings revealed that, for both child-report and parent proxy-report of quality of life, the Barth Syndrome sample reported significantly lower scores on the total score and all domains of quality of life (see Table 3).

### Comparison of Behavior Problems, Adaptive Functioning, and Social Support in Youth with Barth Syndrome and Healthy Controls

Scores from the CBCL, ALS, ABS-S2, and PPSS for the Barth Syndrome sample and the healthy control sample are presented in Table 4. Independent-sample *t*-tests were conducted to examine between-group differences on these measures. Results revealed significant group differences on the CBCL total score, the CBCL Internalizing scale, the CBCL Externalizing scale, and the CBCL Social Problems subscale, such that scores for the Barth Syndrome sample were significantly higher (indicating more problems) than those for the healthy control sample. Significant group differences were also found on the ALS, with boys with Barth Syndrome reporting significantly higher levels of loneliness compared to healthy controls.

Regarding adaptive functioning, boys with Barth Syndrome evidenced significantly lower scores (indicating worse functioning) compared to the healthy control sample on the ABS-S2 subscales of Independent Functioning (assessing self-care tasks), Physical Development (assessing sensory and motor abilities), Economic (assessing tasks like managing money, budgeting, and shopping), Language (assessing receptive and expressive abilities), Numbers

and Time (assessing mathematical competencies), Prevocational/vocational Activity (assessing school- or job-related skills), Self-Direction (assessing the degree to which one maintains an active lifestyle), Responsibility (assessing dependability with tasks such as being punctual and maintaining self-control), and Socialization (assessing ability to interact with others). No significant group differences were found on the PPSS.

### Comparison of Parental and Family Functioning in Youth with Barth Syndrome and Healthy Controls

Table 5 presents scores obtained from parent ratings on the BSI Global Stress Index, PIP, and child ratings on the SRQ for the Barth Syndrome sample and the healthy control sample. Independent-sample *t*-tests revealed significant group differences for ratings of parental stress associated with caring for a child with a medical illness (as assessed by the PIP), as well as general parental distress (e.g., anxiety and depressive symptoms; as assessed by the BSI). Specifically, parents of boys with Barth Syndrome reported significantly higher scores than parents of healthy children (indicating more stress and distress) on the PIP-F score, the PIP-D score, and the BSI Global Severity Index. No significant group differences emerged on any of the SRQ subscale scores ($p > .05$; See Table 5).

### Intellectual and School Functioning Among Youth with Barth Syndrome

The mean standard score on the PPVT-III for youth with Barth Syndrome was 106.34 (*SD* = 13.07) and the median was 110. Scores on the PPVT-III ranged from 84 to 128. Seventy-four percent of parents reported that their son received school accommodations and these accommodations included classroom seating changes (52%), rest periods (48%), schedule adjustments (39%), special diets (22%), note takers (22%), extra books for home use (22%), alternative assignments (17%), medication administration (17%), extra tutorials (13%), use of tape recorders (9%), and peer mentors (9%). Twenty-six percent of parents reported that their child was being monitored by a school psychologist, and 22% reported that their child had close contact with a school guidance counselor. Regarding communication, 65% of parents reported that they regularly communicated with their child's teachers, but parents reported that only 13% of their child's teachers regularly communicated with their child's heath care team. School attendance rates during the last 12 months and during the worst episode of illness are reported in Table 6.

## Discussion

This pilot study examined psychosocial functioning and parental distress in boys with Barth Syndrome, a rare genetic disorder associated with a multitude of physical abnormalities and oftentimes, early mortality. Consistent with our expectations and findings in children with other chronic health conditions (Hysing et al., 2007; Varni et al., 2003, Varni et al., 2004; Williams et al., 2005), findings generally indicated that boys with Barth Syndrome experience lower quality of life across all domains when compared to healthy children, including psychosocial health, physical, emotional, social, and school functioning. In addition, results indicate that boys with Barth Syndrome experience significantly lower quality of life than boys with cardiac disease alone (Varni et al., 2007), further emphasizing the impact of this illness on psychosocial functioning.

Parents of children with Barth Syndrome reported that their children exhibited some deficits in independent functioning relative to healthy children. Specifically, compared to healthy peers, boys with Barth Syndrome were rated by their parents as having decreased abilities in areas such as financial management, independent shopping, and prevocational/vocational activity (i.e., school- or job-related skills). One way of understanding these differences is that there may be fewer opportunities to engage in activities that help develop adaptive skills, due

to the higher frequency of hospitalizations and medical care commonly seen in boys with Barth Syndrome. Further, parents of boys with Barth Syndrome may be more restrictive of their children's activities because of their often realistic concerns about the effect on their child's health. Consistent with this, and similar to families of children with other chronic health conditions (e.g., Montagnino & Mauricio, 2004; Silver et al., 1995), parents of boys with Barth Syndrome reported significant distress associated with caring for their son with a chronic illness.

Interestingly, boys with Barth Syndrome rated their own psychosocial functioning in a similar manner as their parents. This pattern was also exhibited in social and behavioral domains, as parents of youth with Barth Syndrome reported significantly higher levels of behavioral and social problems on the CBCL than healthy controls; whereas, the youth themselves reported significantly higher levels of loneliness than controls on the ALS. This parent-child congruency is not typically seen in families of children with chronic illnesses, as parents typically report more observed problems in their child than the child him/herself (e.g., Bastiaansen et al., 2004; Varni et al., 2003; Williams et al., 2005). The congruency between parent and child reports could be attributed to the severity of the illness and the resulting increase in communication and information sharing between the parent and child. This congruency may also be related to the number of objective illness indicators such as the high frequency of doctor's office visits, complex medication regimen, and family appraisal of the illness. However, further research in this area would be necessary before making any concrete conclusions.

Academically, boys with Barth Syndrome demonstrated receptive vocabulary abilities similar to youth without a chronic illness. This is in slight contrast to Mazzocco et al. (2007) who found that scores on the Wechsler Abbreviated Scale of Intelligence were significantly lower than healthy controls (effect size = −0.91) for 12 boys with Barth Syndrome (age ≥ 6 years). This finding may be a function of a sampling bias in that more highly functioning boys with Barth Syndrome were able to attend the conference and thereby participated in this research. In addition, many parents in the pilot study endorsed the need for school-based accommodations to assist their child academically.

Although this report marks the most substantial psychosocial investigation into Barth Syndrome published to date, it is important to note that these findings are preliminary in nature and several study limitations exist. First, the relatively small sample size limited our ability to conduct higher-level statistical analyses (e.g., interactional analyses) that could allow for the examination of the effects of variables such as demographics (e.g., age, country of origin) and physical health (e.g., medical severity). However, this study included a relatively large percentage of the population of interest, given the extremely small number of identified patients with Barth Syndrome worldwide (Kelley, 2007). Second, we included subjects of a broad age range. Some study measures are not normed for older individuals and developmental functioning likely differs as a function of age. On balance, the majority of participants were 18 years or younger, and those who were older still were extremely reliant on their families for support (e.g., living at home, required financial assistance). Given the preliminary nature of this study, we felt it most informative to include a broad age range that may inform future research. Third, with the exception of the PPVT-III, all of the assessments used in this study were self-report (or parent-proxy-report) measures. Therefore, the results could potentially have been affected by differences in response patterns. In order to provide a more comprehensive assessment of boys with Barth Syndrome, future studies should incorporate methods of assessment from sources outside of the patient and family (e.g., teacher report, clinician-report, and objective data). Finally, this study represents a cross-sectional snapshot into the psychosocial functioning of boys with Barth Syndrome. There are numerous other variables of interest to examine such as how varied illness factors (e.g., severity, amount of

ongoing medical care) interact with adjustment. Further, it will be necessary to examine psychosocial issues in this population in a prospective manner. It is quite conceivable, for example, that psychosocial effects on children and their families may differ at various developmental epochs, something that we were not able to investigate statistically given the small sample. However, we would hypothesize that the initial period of diagnosis is quite stressful given the realistic mortality concerns and demands placed on the parents. Similarly, adolescence is likely to be a difficult period as, similar to other chronic illnesses (e.g., Kovacs, Goldston, Obrosky, & Iyengar, 1992), adherence to medical regimens may be less than optimal.

## Implications for Practice

The present findings have implications for academic functioning and psychosocial intervention for the child and family. Regarding the former, given the benefits of including youth with disabilities in general education settings (Fisher & Meyer, 2002), boys with Barth Syndrome would likely benefit from such environments where teachers and support staff (i.e., special education teachers or paraprofessionals) can accommodate their unique academic and physical needs. For example, teachers can provide opportunities for boys with Barth Syndrome to participate in physical activities and games with typically developing peers while adhering to their regimen and restrictions. Because many parents struggle to navigate the vicissitudes of securing appropriate educational services for their child with a disability (Kluth, Biklen, English-Sand, & Smukler, 2007), it would be helpful for school professionals (i.e., general and special education teachers and school administrators), health professionals (i.e., school nurses and other medical specialists), and families of boys with Barth Syndrome to work as a transdisciplinary collaborative team. This would facilitate effective communication and allow them to share strategies for achieving personal and academic gains. As reported by many parents in this study, ongoing and regular communication with such professionals is critical for both the academic and psychosocial success of a boy with Barth Syndrome.

Given the significant psychosocial issues reported by youth with Barth Syndrome and their parents, it seems likely that families would benefit from consultation with a psychologist or mental health professional in order to learn adaptive ways to manage their increased responsibilities and monitor psychosocial functioning in their child with Barth Syndrome. Specifically, parents may benefit from learning coping skills to manage their own personal distress while attempting to balance the burden of helping their child and maintaining a healthy lifestyle. Coping skills such as relaxation techniques (i.e., progressive muscle relaxation, deep breathing, guided imagery) could help parents to better manage their own levels of anxiety. In addition, parents may benefit from group intervention programs, such as the one implemented by Dellve and colleagues (2006), for parents of children with rare diseases. This program focuses on empowering parents by enhancing a range of competencies that are believed to impact quality of life. Families travel to a designated location for a 5-day long intensive program that includes sessions for parents, the affected child, their sibling(s), and the entire family. For parents, specific areas of focus include education about the disease, evidence-based treatment approaches to disease management, available social services and related resources. For the affected child and his/her siblings, there are discussions about disease management, the experience of having a condition (or an affected sibling), and other pragmatic issues (e.g., negotiating peer concerns). In addition to the practical information conveyed, interventions such as this provide emotional support and enable parents to share information, thoughts, and feelings with others who have similar situations.

Finally, some parents and other caregivers (e.g., teachers) for boys with Barth Syndrome may benefit from professional assistance in determining appropriate opportunities for the child to participate in activities independently. By identifying not only specific activities that should be avoided, but also those which are allowable, unnecessary restrictions made by uninformed

but well-intended adults will be minimized. However, it is imperative that clinicians provide this information as a component of ongoing disease management education, as this information can ultimately improve both the child's and parents' quality of life.

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**Table 1**

Descriptive statistics and bivariate analyses of quality of life

| | Healthy control sample [a] (*n* = 22) | | Cardiac disease sample [b] (*n* = 344) | | Barth Syndrome sample [c] (*n* = 34) | | Group comparison |
|---|---|---|---|---|---|---|---|
| | *M* | *SD* | *M* | *SD* | *M* | *SD* | |
| PedsQL | | | | | | | |
| Total score | 86.47 | 6.32 | 77.47 | 14.51 | 55.22 | 15.94 | a > b > c |
| Physical functioning | 92.97 | 5.18 | 82.28 | 15.68 | 45.39 | 17.96 | a > b > c |
| Psychosocial health | 82.44 | 10.51 | 74.88 | 16.10 | 59.44 | 17.12 | a > b > c |
| Emotional functioning | 75.38 | 15.34 | 73.78 | 20.38 | 61.05 | 17.61 | a = b > c |
| Social functioning | 90.77 | 9.76 | 78.74 | 19.52 | 60.25 | 21.30 | a > b > c |
| School functioning | 81.15 | 12.93 | 72.09 | 19.01 | 56.84 | 21.74 | a > b > c |
| PedsQL Parent proxy | | | | | | | |
| Total score | 94.37 | 4.92 | 79.44 | 16.50 | 53.40 | 21.11 | a > b > c |
| Physical functioning | 97.47 | 6.38 | 83.11 | 18.73 | 39.78 | 27.48 | a > b > c |
| Psychosocial health | 93.33 | 6.19 | 77.36 | 17.27 | 57.43 | 19.12 | a > b > c |
| Emotional functioning | 88.81 | 10.59 | 74.69 | 20.45 | 59.48 | 21.56 | a > b > c |
| Social functioning | 97.14 | 4.89 | 82.52 | 20.11 | 58.28 | 26.80 | a > b > c |
| School functioning | 94.05 | 9.70 | 73.09 | 20.35 | 55.00 | 21.26 | a > b > c |

*Note.* PedsQL = Pediatric Quality of Life Inventory; “>” 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.

## Table 2

Independent sample t-tests comparing Barth Syndrome sample and healthy controls

| | *t*-test | | Effect size (CI) | *CI for means* |
|---|---|---|---|---|
| | *t* | *p* | | |
| PedsQL (n = 33) | | | | |
| Total score | −7.31 | < .01 | −2.36 (−3.05 to −1.67) | −40.11 to −22.39 |
| Physical functioning | −10.85 | < .01 | −3.26 (−4.06 to −2.45) | −56.66 to −38.49 |
| Psychosocial health | −4.62 | < .01 | −1.52 (−2.13 to −0.92) | −33.18 to −12.80 |
| Emotional functioning | −2.38 | < .05 | −0.84 (−1.40 to −0.28) | −26.35 to −2.32 |
| Social functioning | −4.83 | < .01 | −1.70 (−2.32 to −1.08) | −41.73 to −19.31 |
| School functioning | −3.61 | < .01 | −1.27 (−1.86 to −0.39) | −36.87 to −11.76 |
| PedsQL Parent proxy (n = 50) | | | | |
| Total score | −8.66 | < .01 | −2.41 (−3.10 to −1.71) | −50.77 to −31.16 |
| Physical functioning | −10.36 | < .01 | −2.60 (−3.32 to −1.88) | −69.08 to −46.29 |
| Psychosocial health | −8.69 | < .01 | −2.28 (−2.96 to −1.60) | −44.36 to −27.45 |
| Emotional functioning | −6.34 | < .01 | −1.60 (−2.21 to −0.99) | −39.60 to −19.05 |
| Social functioning | −7.64 | < .01 | −1.81 (−2.44 to −1.18) | −50.80 to −26.93 |
| School functioning | −8.09 | < .01 | −2.18 (−2.85 to −1.51) | −49.23 to −28.86 |

*Note*. PedsQL = Pediatric Quality of Life Inventory

## Table 3

One sample t-tests comparing Barth Syndrome sample and cardiac disease sample

| | ***t*-test** | | **Effect size (CI)** | ***CI for means*** |
|---|---|---|---|---|
| | *t* | *p* | | |
| PedsQL (n = 19) | | | | |
| Total score | −6.24 | < .01 | −1.52 (−2.00 to −1.03) | −31.84 to −15.62 |
| Physical functioning | −11.41 | < .01 | −2.32 (−2.83 to −1.82) | −46.68 to −32.11 |
| Psychosocial health | −3.92 | < .01 | −0.95 (−1.43 to −0.48) | −25.19 to −7.51 |
| Emotional functioning | −2.91 | < .05 | −0.63 (−1.10 to −0.16) | −21.37 to −3.41 |
| Social functioning | −4.38 | < .01 | −0.94 (−1.41 to −0.47) | −30.06 to −10.58 |
| School functioning | −3.15 | < .01 | −0.79 (−1.26 to −0.32) | −27.15 to −5.36 |
| PedsQL Parent proxy (n = 28) | | | | |
| Total score | −6.17 | < .01 | −1.54 (−1.94 to −1.14) | −37.16 to −18.33 |
| Physical functioning | −9.12 | < .01 | −2.22 (−2.63 to −1.80) | −56.09 to −35.38 |
| Psychosocial health | −5.39 | < .01 | −1.14 (−1.54 to −0.75) | −29.15 to −12.96 |
| Emotional functioning | −3.91 | < .01 | −0.74 (−1.13 to −0.35) | −24.31 to −7.57 |
| Social functioning | −5.23 | < .01 | −1.17 (−1.56 to −0.78) | −35.83 to −15.64 |
| School functioning | −4.18 | < .01 | −0.88 (−1.27 to −0.49) | −28.04 to −9.45 |

*Note*. PedsQL = Pediatric Quality of Life Inventory

**Table 4**

Descriptive statistics and bivariate analyses of behavior problems and adaptive functioning

| | Healthy control sample (*n* = 21) | | Barth Syndrome sample (*n* = 29) | | *t*-test | | Barth Syndrome Effect size (CI) | 30 CI for means |
|---|---|---|---|---|---|---|---|---|
| | *M* | *SD* | *M* | *SD* | *t* | *p* | | |
| CBCL | | | | | | | | |
| Total score | 6.64 | 6.53 | 27.45 | 15.58 | 4.54 | < .01 | 1.62 (.98 to 2.27) | 11.32 to 30.31 |
| Internalizing | 1.94 | 2.70 | 7.19 | 4.97 | 3.74 | < .01 | 1.24 (.63 to 1.85) | 1.40 to 2.34 |
| Externalizing | 2.33 | 2.66 | 7.00 | 4.19 | 3.89 | < .01 | 1.27 (.65 to 1.88) | 2.22 to 7.12 |
| Social Problems | .37 | .68 | 2.63 | 2.45 | 3.57 | < .01 | 1.16 (.55 to 1.76) | .92 to 3.59 |
| ALS Total Score | 23.27 | 4.80 | 32.74 | 10.46 | 2.82 | < .01 | 1.09 (.49 to 1.69) | 2.59 to 16.34 |
| PPSS Total Score | 5.92 | 1.88 | 6.00 | 2.29 | .11 | .92 | .04 (−0.52 to 0.60) | −1.53 to 1.70 |
| ABS-S2 | | | | | | | | |
| Independent functioning | 101.95 | 12.82 | 77.21 | 27.95 | −4.20 | < .01 | −1.06 (−1.66 to −0.46) | −36.65 to −12.84 |
| Physical development | 24.00 | .00 | 21.07 | 3.61 | −4.37 | < .01 | −1.05 (−1.64 to −0.45) | −4.31 to −1.56 |
| Economic | 14.62 | 7.55 | 9.18 | 8.39 | −2.34 | .02 | −0.67 (−1.24 to −0.09) | −10.11 to −0.77 |
| Language | 40.10 | 3.56 | 33.17 | 10.70 | −3.24 | < .01 | −0.80 (−1.39 to −0.22) | −11.25 to −2.60 |
| Numbers and time | 13.00 | 1.79 | 9.96 | 4.69 | −3.09 | < .01 | −0.80 (−1.38 to −0.21) | −5.03 to −1.04 |
| Prevocational / vocational | 10.38 | .67 | 6.41 | 3.27 | −6.15 | < .01 | −1.54 (−2.18 to −0.90) | −5.30 to −2.65 |

*Note.* CBCL = Child Behavior Checklist; ASL = Asher Loneliness Scale; ABS-S2 = Adaptive Behavior Scale-School, Second Edition; CI = 95% Confidence Interval

**Table 5**

Descriptive statistics and bivariate analyses of parental and family functioning

| | Healthy Control sample (*n* = 19) | | Barth Syndrome sample (*n* = 22) | | *t*-test | | Effect size (CI) | CI for means |
|---|---|---|---|---|---|---|---|---|
| | *M* | *SD* | *M* | *SD* | *t* | *p* | | |
| BSI Global Distress | 6.75 | 8.86 | 26.43 | 17.36 | 4.11 | < .01 | 1.37 (.69 to 2.05) | 9.81 to 29.56 |
| PIP | | | | | | | | |
| PIP-F | 50.79 | 6.92 | 103.09 | 22.90 | 10.19 | < .01 | 2.94 (2.06 to 3.82) | 41.74 to 62.87 |
| PIP-D | 47.95 | 7.39 | 97.45 | 22.88 | 9.18 | < .01 | 2.77 (1.91 to 3.63) | 38.36 to 60.65 |
| SRQ child-report | | | | | | | | |
| Warmth/Closeness | 2.39 | 1.04 | 2.21 | .65 | −0.51 | .62 | −0.21 (−0.82 to 0.41) | −0.97 to .60 |
| Conflict | 2.18 | .96 | 1.76 | .98 | −1.13 | .27 | −0.42 (−1.04 to 0.20) | −1.20 to .35 |
| Rivalry | 1.85 | .43 | 2.06 | .49 | 1.16 | .26 | .44 (−0.18 to 1.07) | −0.15 to .58 |

*Note.* BSI = Brief Symptom Inventory; PIP = Pediatric Inventory for Parents; PIP-F = Pediatric Inventory for Parents-Frequency; PIP-D = Pediatric Inventory for Parents-Difficulty; SRQ = Sibling Relationship Questionnaire; CI = 95% Confidence Interval

**Table 6**

School Attendance Rates of Youth with Barth Syndrome

| | During Last 12 Months | | During Worst Episode | |
|---|---|---|---|---|
| | *Frequency* | *Percent* | *Frequency* | *Percent* |
| None or only odd days off | 5 | 26 | 3 | 16 |
| Attending at least 85% of the time | 11 | 58 | 6 | 32 |
| Between 50 and 85% of the time | 1 | 5 | 3 | 16 |
| Less than 50% attendance | 1 | 5 | 4 | 21 |
| Has not attended school for entire academic term | 1 | 5 | 3 | 16 |