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Original Article

A New Perspective on the Quality of Life of Children with Glycogen Storage Diseases

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Conflict of Interest
The authors have no financial conflicts of interest.

ABSTRACT

Purpose: This study aimed to assess the quality of life (QoL) of children with glycogen storage disease (GSD) and their parents and to determine the impact of myopathies.
Methods: A prospective case-control study was conducted at the Cairo University Children's Hospital and National Liver Institute, Menoufia University. A promising new style of questionnaire called the Stark Quality of Life Questionnaire was used to assess the quality of life.
Results: Fifty-two children diagnosed with GSD (cases) and 55 age- and sex-matched healthy children (controls) were included. A statistically significant difference was found between cases and controls regarding food intake; mental behavior parameters such as mood, energy, and social contact; and physical behavior parameters such as running and tying shoelaces. Children with myopathies had significantly lower QoL scores in most of the parameters.
Conclusion: GSDs alter children and their parents' mental and physical abilities. Lower QoL scores were detected in children with both skeletal myopathy and cardiomyopathy, but the difference was not statistically significant when compared with the children without myopathies.

Keywords: Quality of life; Glycogen storage diseases; Muscular diseases

INTRODUCTION

Glycogen storage diseases (GSDs) are metabolic disorders caused by enzyme deficiencies that affect glycogen synthesis, glycogen breakdown, or glycolysis within the muscles, liver, heart, and other cell types [1].

Hypertrophic cardiomyopathy is an autosomal dominant disorder associated with increased morbidity and premature mortality. It is diagnosed based on an increased cardiac mass [2].

The main clinical symptoms of hepatic disease are hypoglycemia and hepatomegaly [3]. A characteristic feature of muscle involvement in patients with GSDs is progressive myopathy [4]. Metabolic acidosis with hypercholesterolemia and hyperlipidemia has also been reported

[5]. GSDs are a group of heterogeneous genetic diseases; therefore, each type has a specific clinical presentation [6].

A study on children with GSD III concluded that myopathic changes are common in children with GSD III. Myopathic changes tend to occur in older patients and are associated with higher creatine phosphokinase levels [7].

Measurement of quality of life (QoL) has become important in applied medicine. Randomized controlled trials and observational studies of various disease conditions have increasingly included QoL assessment [8]. In modern medicine, the targets of medical teams are directed not only to improve the physical symptoms of the disease but also to help improve the overall QoL. In GSDs, QoL can be affected by the disease itself and its complications due to the strict diet regimen and prescribed medications.

This study aimed to assess the QoL of children with GSDs and their parents. We also aimed to compare the QoL of patients with and without skeletal myopathies and hypertrophic cardiomyopathy. To the best of our knowledge, this is the first study to assess these parameters in a group of metabolic disorders.

MATERIALS AND METHODS

Participants

A prospective case-control study was conducted on patients attending the Cairo University Children's Hospital (Cairo, Egypt) and National Liver Institute (Menoufia, Egypt) from May 2018 to November 2019. Fifty-two children diagnosed with GSD, some of whom were newly diagnosed in this period, plus cases diagnosed before and following up in our clinics, with or without muscle and cardiac affection due to the disease, were included in the study. The sample size was calculated using G*Power C software version 3.1.7 (Institute of Experimental Psychology, Heinrich Heine University, Dusseldorf, Germany), assuming alpha-error=0.05 (two tails) and beta-error=0.1 (power=90%). The ages of the included patients ranged from 1 to 18 years. Patients with motor or mental disabilities, other chronic illnesses, and those with cardiac or musculoskeletal abnormalities due to causes other than GSD were excluded from the study. The entire study group was classified into three groups according to the presence or absence of cardiac and skeletal myopathies (**Table 1**).

1 - Group A: represents children with GSDs that are not complicated with skeletal myopathy or hypertrophic cardiomyopathy.
2 - Group B: represents children with GSDs that are complicated with either isolated hypertrophic cardiomyopathy or isolated skeletal myopathy.
3 - Group C: represents children with GSDs that are complicated with both skeletal myopathy and hypertrophic cardiomyopathy.

Data on GSDs were collected and analyzed.

All patients (children diagnosed with GSD) and controls (healthy age- and sex-matched children) were interviewed at least once, and one of their parents completed the Stark QoL Questionnaire [9].

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Table 1. Frequency of study groups and subtypes of GSDs

Study groups and GSD subtypes	Number	%
Study groups		
No skeletal myopathy or cardiomyopathy (group A)	28	53.8
Either skeletal myopathy or cardiomyopathy (group B)	12	23.1
Both skeletal myopathy or cardiomyopathy (group C)	12	23.1
GSD types		
Unknown	15	14.0
Type I	5	4.7
Type Ib	1	0.9
Type III	14	13.1
Type IIIa	12	11.2
Type IIIb	3	2.8
Type IV	1	0.9
Type IX	1	0.9

GSD: glycogen storage disease.

Materials

Each case (children diagnosed with GSD) in the study was subjected to the following: Full history was noted, including assessment of hepatic symptoms and signs, recurrent chest infection, vomiting, bleeding tendency, diarrhea, fever, abdominal pain, convulsions, encephalopathy, lower limb edema, behavioral changes, abdominal enlargement, and hypoglycemia. For cardiac symptoms and signs, shortness of breath, chest pain, palpitations, and fainting were assessed.

A thorough clinical examination, including doll facies, short stature, failure to thrive, hepatomegaly, and murmur, was conducted. Complete family pedigree construction. Investigations in the form of liver function tests, complete blood count, lipid profile, uric acid, creatinine, and prothrombin time. Imaging in the form of abdominal ultrasound, chest X-ray, ECG, 2D, and color Doppler echocardiography in patients with hypertrophic cardiomyopathy.

Every child in the control group (healthy age- and sex-matched children) was subjected to history-taking, including family history and history of any hepatic affection. Blood tests in the control group included complete blood count, liver enzymes (aspartate transaminase and alanine transaminase), and prothrombin time.

The Stark QoL Questionnaire [9] was used to assess both the mental and physical components of QoL. This questionnaire had not been used with Egyptian children previously, but we preferred to use it as it includes pictures and contains a minimum of words, making it suitable for this age group. It is called Stark because the pictures were drawn by a German artist named H.P. Stark, and formal permission was obtained through personal communication (Stark H.P., personal communication, January 16, 2020) [10]. It is considered a promising new style in measuring QoL and international research as it is a short and easy-to-apply questionnaire. The Stark QoL comprises 16 pictures representing different mood statuses, energy, social contact, and various physical activities. The content of the items was transferred into the pictures, leaving only short text elements in between. Respondents had to choose a symbol ("--," "-," "0," "+," "++") near the picture describing how well they could perform a certain task. Since approximately 10% of the world's population is still illiterate, it was an excellent advantage that illiterate respondents would be able to fill it out.

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Regarding GSDs types, the majority were GSD type III, followed by type IIIa, type I, and type IIIb, versus a minority of patients with an unknown type of GSD (enzyme assay not performed), but most probably they are type III due to their phenotypic picture, and type III is the most common in our population (**Table 1**). The results of a study conducted on children with GSD III in 2009 suggested allelic and phenotypic heterogeneity of GSD III in our country [11]. We used different types of GSD in the study (I, IIIa, IIIb, unidentified type) according to the results of the enzyme assay, if done. Patients with GSD type IIIa are thought to have cardiac and skeletal affections. However, clinically, some of these patients may have cardiac or skeletal affection during the early course of the disease, or the cardiac or muscle affection may appear later. Moreover, some patients who cannot afford the enzyme assay may have a clinical cardiac or skeletal affection, with no type identified.

Procedure

*1. Application of the Stark QoL Questionnaire (**Fig. 1**)*

The first item measures the mood and consists of five smiley faces; at one end is a very happy face, and at the other end a very sad one. The patients were asked to check the one that most correlated with their mood. The second item measures energy and presents three pictures of a walking person: on the left-hand side, the walker is full of energy, and on the right, he seems to be walking as if depressed. The third item measures social contact, and three pictures show a group of five persons each, one white and four gray. The white person symbolizes the patient himself, and the gray one is a possible peer group. On one end, the white person is standing in the middle of the group and on the other end, alone. Together, these three items constitute the mental component.

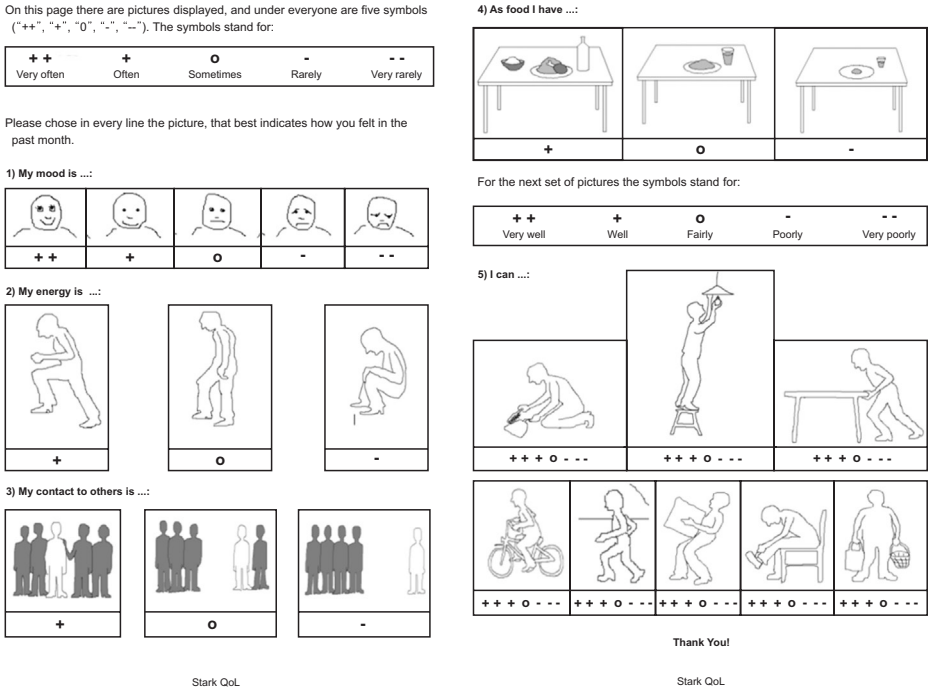

Fig. 1. The Stark Quality of Life questionnaire (quoted from: A new questionnaire for measuring the quality of life - the Stark QoL, 2015) (Stark H.P., personal communication, January 16, 2020).

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On the second page, eight items measuring physical activities are presented. The pictures show activities including carrying a shopping basket, moving a table, tying shoelaces, etc. The text reads "I can," and "++" stands for "very well," "+" for "well," "0" for "fairly," "-" for "poorly", and "--" for very poorly. The patients were asked to indicate how easily they could perform the activity displayed in each picture. These items constitute the physical components. All the items in the present analysis were coded between 0 and 100. Scales were calculated as the mean of the items, with high values indicating good QoL. The collected data were organized, tabulated, and statistically analyzed.

Statistical analysis

Data were collected and coded to facilitate data management and entered into Microsoft Access. Data analysis was performed using SPSS software version 18 (IBM Co., Armonk, NY, USA) in Windows 7. Simple descriptive analyses in the form of numbers and percentages for qualitative data, arithmetic means as central tendency measurement, and standard deviations as a measure of dispersion for quantitative parametric data were performed. Quantitative data included in the study was first tested for normality using the one-sample Kolmogorov–Smirnov test in each study group, and inferential statistical tests were selected.

For quantitative parametric data, an independent Student's *t*-test was used to compare measures of two independent groups of quantitative data. One-way analysis of variance was used to compare more than two independent groups of quantitative data with Bonferroni post-hoc test to assess the significance between the two groups. For quantitative nonparametric data, the Kruskal–Wallis test was used to compare more than two independent groups. The Mann–Whitney test was used to compare two independent groups.

For qualitative data, a chi-square test was used to compare two or more qualitative groups. The probability p-value≤0.05 was considered the cut-off value for statistical significance.

Ethical approval

All procedures performed in studies involving human participants followed the ethical standards of the Faculty of Medicine's Research Ethical Committee and the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This study was approved by the Institution Review Board of the National Liver Institute, Menoufiya University (00304/2022). Informed consent was obtained from all the participants included in the study.

RESULTS

Descriptive results

We enrolled a convenience sample of 52 children and adolescents (aged 1–18 years) with confirmed GSDs, with and without hypertrophic cardiomyopathy and skeletal myopathy, and 55 age- and sex-matched healthy controls. In addition, we enrolled 52 parents for assessment of QoL, either the child's father or mother, and 55 sex-matched parents of healthy children (controls). Thirteen patients with GSDs had confirmed hypertrophic cardiomyopathy, 23 had skeletal myopathy, and 12 had combined cardiac and skeletal myopathic complications. Among classified groups of patients, group A, which represents children with GSDs that are not complicated with skeletal myopathy or hypertrophic cardiomyopathy, constituted 53.8% (28 patients) of the entire study group. Conversly, group B, which represents children with

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Table 2. Comparisons of demographic characters in different study groups

Variable	Case (n=52)	Control (n=55)	*p*-value
Age (yr)	7.4±4.3	8.1±2.6	0.4
Sex			0.2
Male	31 (59.6)	26 (47.3)	
Female	21 (40.4)	29 (52.7)	
Sex of parents			0.3
Male	6 (11.5)	11 (20.0)	
Female	46 (88.5)	44 (80.0)	

Values are presented as mean±standard deviation or number (%).

GSDs that are complicated with either isolated hypertrophic cardiomyopathy or isolated skeletal myopathy, constituted 1.9% and 21.2% (12 patients) of the entire study group, respectively. Lastly, group C represents children with GSDs complicated by both skeletal myopathy and hypertrophic cardiomyopathy, constituting 23.1% (12 patients) of the entire study group.

The mean age of patients was 7.4±4.3 years; among them, 59.6% were males. There was no statistically significant difference between cases and controls with regard to age, sex, and sex of parents, which indicated proper matching between both the groups (**Table 2**).

Among the patients, 26.9% had a family history of other GSD patient(s), and 86.5% had a positive history of consanguineous marriage. The mean body weight was 25.2±13.8 kg and mean height was 108.9±22.9 cm. Most patients were below the 50th percentile for weight and height.

Cases complicated with skeletal myopathy represented 44.2% of cases, and 25% were complicated with hypertrophic cardiomyopathy. Among hypertrophic cardiomyopathy cases, 13.5% had a positive family history.

With regard to GSDs type, 14 patients (13.1%) had GSD type III, followed by 12 patients (11.2%) type IIIa, 5 patients (4.7%) with type I, 3 patients (2.8%) with type IIIb, and 15 patients (14%) patients with an unknown type of GSD (did not undergo enzyme assay).

With regard to patients with cardiomyopathy, the mean interventricular septal thickness was (1.26±0.71) with only 7.7% of cases having arrhythmia.

For the treatment of GSD, cases compliant to uncooked corn starch consumption were 37 (71.2%) patients, but with interruption of sleep to be strict for treatment. All patients were consuming frequent meals rich in complex carbohydrates, with variable restrictions in lactose or fructose intake, and were consuming uncooked cornstarch at least once a day. Drugs used were collected as medications. The patient groups were administered hepatic medications according to their clinical condition. Most of them were on “Ursodeoxycholic acid,” and some were taking “Allopurinol” for hyperuricemia. Some patients were taking lipid-lowering agents such as “Statins.” Some were on “Omega 3” and vitamins, and six patients with hypertrophic cardiomyopathy were taking cardiac medications in the form of propranolol.

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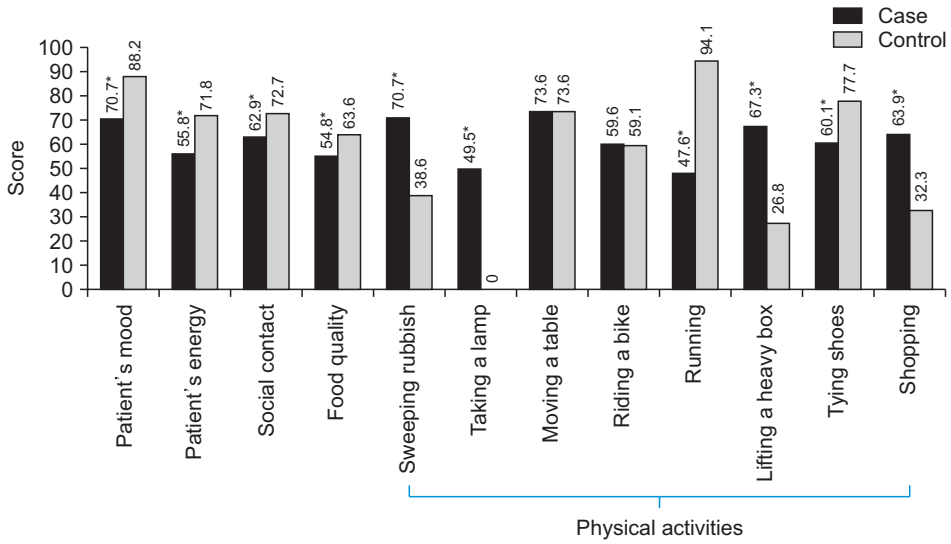

Fig. 2. Comparisons of quality of life score of children with glycogen storage disease and healthy controls in different study groups. *Statistically significant difference with $p<0.05$.

Analytical (comparative) results

1. The Stark QoL Questionnaire of children with GSDs versus control group

Statistical significance was set with a $p<0.05$. Mental components such as mood, energy and social contact were statistically low in the GSD group than in the control group ($p=0.002$, <0.001 and <0.001, respectively). Moreover, statistically significant differences ($p<0.05$) for food intake and some items that constitute physical components, such as running and tying shoelaces, were statistically low in the GSD group than in the control group ($p=0.02$, <0.001 and 0.005, respectively). The statistically significant differences in other items that constitute physical components, including sweeping rubbish, taking a lamp, lifting a heavy box, and shopping, were statistically higher in the GSD group than in the control group ($p<0.001$, <0.001, and <0.001, respectively). On the other hand, there was no statistically significant difference with regards to moving a table, riding a bike, or total score ($p>0.05$) (**Fig. 2**).

2. The Stark QoL Questionnaire of parents of children with GSDs versus control group

There was a statistically significant difference in mood ($p<0.05$). Food intake and some items that constitute physical components, such as running and tying shoelaces, were statistically lower in the GSD group than in the control group ($p<0.001$, <0.001, and <0.001, respectively). In addition, statistically significant differences in other items that constitute physical components, such as taking a lamp and riding a bike, were higher in the GSD group than in the control group ($p<0.001$ and <0.001, respectively). However, there was no statistically significant difference for energy, social contact, sweeping rubbish, moving a table, lifting a heavy box, shopping, and total score ($p>0.05$) (**Fig. 3**).

The classification of the entire study group into three groups was performed as follows: patients without any skeletal or cardiac myopathies (group A); patients with only one type of myopathy, either hypertrophic cardiomyopathy or skeletal myopathy (group B); and the third group of patients with both types of complications, hypertrophic cardiomyopathy and skeletal myopathy (group C). There was no statistically significant difference among the three groups with respect to QoL ($p<0.05$). However, patients with both skeletal myopathy and cardiomyopathy (group C) had lower QoL scores in most of the items of mood, energy, and

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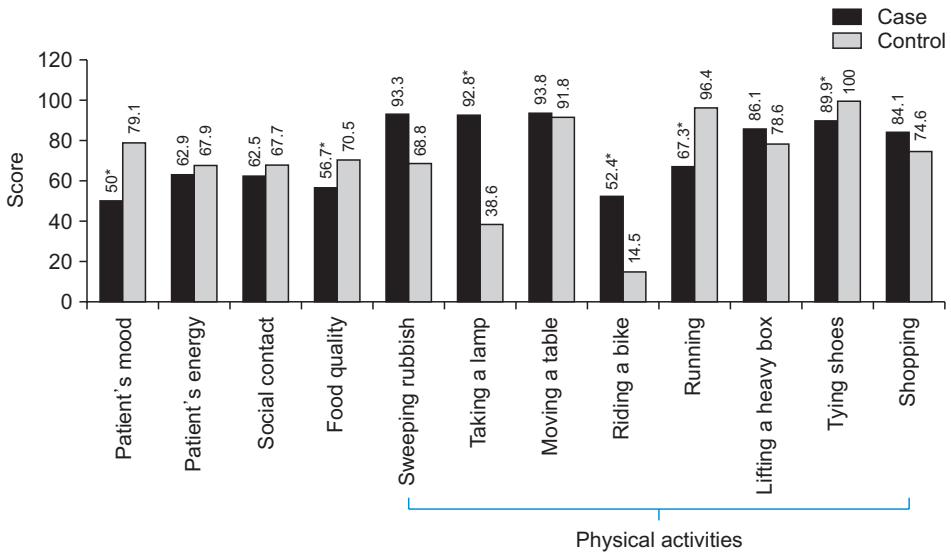

Fig. 3. Comparisons of quality of life score of parents of children with glycogen storage disease and healthy controls in different study groups. *Statistically significant difference with $p<0.05$.

Table 3. Comparisons of quality of life domains score in different groups of children with glycogen storage disease according to the degree of myopathic involvement

Variable	Group A (n=28)	Group B (n=12)	Group C (n=12)	*p*-value
Mood	88.4±19.8	85.4±12.8	62.5±32.8	0.3
Energy	66.9±16.7	66.7±12.3	58.3±19.5	0.6
Social contact	69.6±12.5	70.8±9.7	64.6±16.7	0.9
Food intake	63.4±14.4	62.5±16.9	62.5±16.9	0.9
Sweeping rubbish	66.9±39.1	60.4±36.1	50±43.9	0.4
Taking a lamp	28.6±40.6	22.6±34.4	18.8±26.4	0.8
Moving a table	88.4±24.9	95.8±14.4	56.3±42.8	0.3
Riding a bike	65.2±44.7	50±43.9	62.5±36.2	0.3
Running	80.4±30.7	72.9±36.1	52.1±40.5	0.6
Lifting a heavy	50±41.4	47.9±39.1	50±33.7	0.9
Tying shoes	82.1±33.9	85.4±27.1	60.4±40.5	0.8

Values are presented as mean±standard deviation.
Group A: no skeletal myopathy or cardiomyopathy, Group B: either skeletal myopathy or cardiomyopathy, Group C: both skeletal myopathy and cardiomyopathy.

physical activity, but with no statistically significant difference between this group and the other two groups was found (**Table 3**).

DISCUSSION

There has been a lot of interest in the extent of QoL in people with chronic illnesses, such as GSDs. Doctors and researchers desire to explore how they manage their daily lives [12]. The Stark QoL is an alternative to questionnaires assessing the QoL via worded items. The Stark QoL is a short and direct measure of the two most important dimensions of QoL, and the pictures make the questionnaire easier. In addition, translation into many languages can be easily applied [13].

The study revealed a statistically significant difference with a low mean of three items that constitute mental components such as mood, energy, and social contact in patients with

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GSD compared to controls. The same applies for food intake and some items that constitute physical components, such as running and tying shoelaces. The QoL of patients with GSD I was similar to that reported in patients with type II diabetes. This explains why the drawbacks of the disease and its treatment on the patient's life have a significant impact on general health [14].

A case report done 2 years ago reported a case of acute psychosis in a patient with GSD-Ia. First, they considered opiate use. During treatment, an magnetic resonance imaging scan of the patient's head revealed brain atrophy following significant hypoglycemic insult, thus explaining the organic cause of his psychosis, which is a rare complication of GSD [15]. Thus, it is necessary to explore the organic causes of psychiatric symptoms.

Interestingly, a statistically significant difference with a high mean of other items that constitute physical components included sweeping rubbish, taking a lamp, lifting a heavy box, and shopping, among cases with GSDs compared to controls. This can be attributed to the concept that QoL depends on a person's view of the world and the circumstances in their lives. One way of thinking is to look at the things they can do rather than the things they cannot and to remember that there is always someone worse off than them. Consequently, a high mean for some items constituting physical components was found.

In contrast, a previous study revealed that myopathic symptoms in GSD type IIIa are generally related to muscle wasting later in adult life. Moreover, the inability to debranch glycogen also affects muscle energy metabolism. Six patients aged 17–36 years were studied. They combined anaerobic and aerobic exercises to study skeletal muscle metabolism and exercise tolerance in patients with GSD type IIIa. They found that a part of the skeletal muscle symptoms in GSD type IIIa, that is, weakness and fatigue, may be related to inadequate energy production in the muscle [16].

Regarding the effect of QoL in parents of children with GSDs, we found that there was a statistically significant low mean of mood, food intake, and some items that constitute physical components, such as running and tying shoelaces. Similarly, Storch et al. [17] found a high level of parental stress. There is no apparent relationship between the degree of skeletal and cardiac myopathies and QoL [18].

In the present study, a comparison of the QoL in patients with and without skeletal myopathies and hypertrophic cardiomyopathy showed no statistically significant difference, despite lower scores. This can be explained by the asymptomatic status of most patients with GSDs from the clinical cardiac assessment aspect, except for a very few patients with reported cardiovascular symptoms. Even the reported cardiovascular symptoms are most commonly mild chest pain and arrhythmias, which are temporary symptoms. Annoying cardiac symptoms, such as congestive heart failure and cyanosis, rarely occur in hypertrophic cardiomyopathy. This can be attributed to the fact that myopathic changes tend to occur at an older age [7].

The small number of patients is a limitation of our study, but GSD is one of the rare metabolic diseases that lacks research and publications in the literature, and approaching such a rare disease for research is a strong point of this study.

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GSDs alter the mental and physical abilities of diseased children and their parents. Lower QoL scores were detected in patients with both skeletal myopathy and cardiomyopathy, but the difference was not statistically significant compared to those without myopathy.

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