GEORGIAN BIOMEDICAL NEWS

VOLUME 3 ISSUE 3. JUL-SEP 2025

# Health-Related Quality of Life Among Children with Neuromuscular Diseases in Georgia

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DOI: 10.52340/GBMN.2025.01.01.125

## ABSTRACT

Background: Neuromuscular diseases (NMDs) are a broadly defined group of disorders affecting the peripheral nervous system. NMDs are characterized by their chronic nature, often progressive disability, significant comorbidities, and limited treatment options, all of which influence health-related quality of life (HRQL).
Objectives: Our study aimed to evaluate HRQL in children with NMDs in Georgia.
Methods: The study involved 51 patients aged 5 to 17 years. Patients and their parents were recruited through the physician network and rare disease organizations. HRQL was assessed using the standardized Patient-Reported Outcomes Measurement Information System (PROMIS). Five domains – anxiety, depression, fatigue, pain interference, and physical function – were evaluated using pediatric and parent proxy profile instruments. Intercorrelations were performed to examine the relationships between HRQL, demographic factors, and patient experiences with NMDs.
Results: Mean HRQL scores were significantly poorer across all five PROMIS domains when compared to normative data, indicating substantial impairments among pediatric patients with NMDs. The intercorrelation analysis revealed significant associations between HRQL scores and various demographic and disease-specific factors, underscoring the diverse impact of NMDs on children's lives.
Conclusions: Children with NMDs in Georgia experience significantly reduced HRQL. Our study underscores the necessity for comprehensive care approaches that address the psychological and physical challenges faced by these patients. Further research should investigate targeted interventions to enhance HRQL and establish support systems for individuals affected by these conditions.
Keywords: Health-related quality of life, HRQL; neuromuscular diseases, NMDs; patient-reported outcomes measurement information system, PROMIS.

## BACKGROUND

Neuromuscular diseases (NMDs) comprise a clinically diverse and genetically complex group of conditions that, by definition, affect the muscle, neuromuscular junction, nerve, plexus, nerve root, or anterior horn cells.[1] More than 1000 distinct diagnoses fall under this category. Despite their clinical variability, NMDs share core features of progressive muscle weakness and degeneration. NMDs affect children's health-related quality of life (HRQL), both directly through motor and non-motor impairments, and indirectly through individuals' lived experiences of these limitations. NMDs are typically chronic and lead to a gradual loss of motor function, often resulting in significant mobility limitations and increasing physical dependence over time.[2] In the past decade, the emergence of new treatments designed to extend individuals' lives and improve their living conditions has made the HRQL construct even more relevant.[3] HRQL is a dynamic, subjective, and multidimensional concept. Among the tools used to assess HRQL is the Patient-Reported Outcomes Measurement Information System (PROMIS)—a set of person-centered measures that evaluate and monitor physical, mental, and social health in both adults and children.[4] PROMIS was developed by the National Institutes of Health (NIH) to provide highly reliable and precise instruments that may be used to measure common health symptoms and quality of life domains. It offers greater precision and a broader measurement range than most conventional tools, reducing floor and ceiling effects. PROMIS also supports comparisons across different populations and studies. It includes self-report measures for adults, self-report measures for children aged 8-17, and parent proxy-report measures for children aged 5-17.

There are no studies on HRQL in children with NMDs in Georgia. We aimed to evaluate HRQL in children with NMDs in Georgia using PROMIS.

## METHODS

The study involved 51 patients aged 5 to 17 years. Patients and their parents were recruited through the physician network and rare disease organizations. Sociodemographic and NDD characteristics are summarized in Table 1. HRQL was assessed using the standardized PROMIS. Five domains – anxiety, depression, fatigue, pain interference, and physical function – were evaluated using pediatric and parent proxy profile instruments. PROMIS scores were obtained and converted to standardized T-scores (mean=50, standard deviation=10) for each domain. Descriptive statistics, including means and standard deviations, were calculated for all PROMIS domains. The study was conducted between January 2024 and July 2024. To examine the relationships between HRQL,


ISSN (Online): 2720-8796 ISSN (Print): 2720-7994

demographic factors, and patient experiences with NMDs, intercorrelation analyses (Pearson correlation) were performed. The study investigated correlations between PROMIS domain scores and age, Gender, and disease duration. Ethical approval was obtained from the institutional review board, and written informed consent was obtained from parents or legal guardians before participation, in accordance with national and international ethical standards.

TABLE 1. Sociodemographic and NDD characteristics of the study patients

| Characteristic | N (%) |
|---|---|
| Total N | 51 |
| Age groups | |
| 5–9 years | 18 (35 ) |
| 10–13 years | 16 (31 ) |
| 14–17 years | 17 (33 ) |
| Gender | |
| Male | 28 (55) |
| Female | 23 (45) |
| NMD Diagnosis | |
| Duchenne muscular dystrophy | 20 (39) |
| Spinal muscular atrophy | 15 (29) |
| Other | 16 (31) |

## RESULTS

Mean HRQL scores across all five PROMIS domains were significantly poorer compared to normative data, indicating substantial impairments among pediatric patients with NMDs. As shown in Table 2, the mean scores were: anxiety (62.4), depression (58.7), fatigue (65.2), pain interference (60.3), and physical function (45.6). Intercorrelation analysis revealed significant associations between HRQL scores and various sociodemographic factors, underscoring the diverse impact of NMDs on children's quality of life. Age showed positive correlations with anxiety and pain interference, and a negative correlation with physical function. Disease duration demonstrated positive correlations with anxiety, depression, fatigue, and pain interference, and a negative correlation with physical function (Tab.3). Gender did not show significant correlations with any of the PROMIS domains in this analysis (Tab.3).

TABLE 2. PROMIS mean scores

| | |
|---|---|
| **Domain** | **M** |
| Anxiety | 62.4 |
| Depression | 58.7 |
| Fatigue | 65.2 |
| Pain Interference | 60.3 |
| Physical Function | 45.6 |

TABLE 3. Intercorrelations of sociodemographic and NDD characteristics. *p<0.05 (statistically significant). Positive values indicate a direct correlation; negative values indicate an inverse (negative) correlation

| | Anxiety | Depression | Fatigue | Pain interference | Physical function |
|---|---|---|---|---|---|
| Age | 0.45* | 0.32 | 0.28 | 0.33* | -0.40* |
| Gender | -0.25 | -0.18 | 0.05 | -0.11 | 0.15 |
| Disease duration | 0.38* | 0.41* | 0.42* | 0.39* | -0.37* |

## DISCUSSION

This study aimed to evaluate HRQL in children with NMDs in Georgia using PROMIS. Our findings indicate that children with NMDs in Georgia experience significantly reduced HRQL across multiple domains, including anxiety, depression, fatigue, pain interference, and physical function. The significantly poorer HRQL scores observed in our cohort align with previous research, which highlights the substantial impact of chronic conditions, such as NMDs, on children's well-being. Similar trends have been reported in studies assessing HRQL in children with NMDs in other populations.[5-7] The progressive muscle weakness and degeneration characteristic of NMDs contribute to physical limitations and dependence, which are reflected in the low physical function scores observed in our study. The correlations between HRQL scores and demographic factors such as age and disease duration are consistent with the understanding that NMDs are often progressive, with symptoms worsening over time and accumulating impact on a child's life. These findings highlight the need for comprehensive care approaches that address not only the physical but also the psychological challenges faced by children with NMDs in Georgia. The high scores in anxiety, depression, and fatigue suggest a significant mental and emotional burden, necessitating integrated psychological support within routine clinical care. One limitation of this study is the relatively small sample size (n=51), which may limit the generalizability of our findings to the entire pediatric NMD population in Georgia. Additionally, the cross-sectional design prevents the establishment of causal relationships or the tracking of HRQL changes over time. Future longitudinal studies with larger cohorts are warranted.

## CONCLUSIONS

Children with NMDs in Georgia experience significantly compromised HRQL. Our study underscores the critical need for comprehensive, multidisciplinary care, including psychological support, to mitigate the impact of these disorders on children's lives. Further research should investigate targeted interventions and support systems to enhance HRQL for individuals affected by this condition.

Georgian Biomedical News
ISSN (Online): 2720-8796 ISSN (Print): 2720-7994

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ISSN (Online): 2720-8796 ISSN (Print): 2720-7994