Self- and Parent-Reported Physical and Psychosocial Outcomes for Children with Arthrogryposis

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Research Article

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Abstract

Background

Children with arthrogryposis have impaired motion and strength; however, they develop compensatory strategies to achieve better function than may be anticipated. Impairment measures, such as range of motion and strength, describe physical function, but do not characterize a person’s psychosocial function, or their ability to perform activities and participate in life roles. The Patient Reported Outcome Measurement Information System (PROMIS®) measures domains of function from the perspective of people with a chronic disease, compared to a reference population. We hypothesized that PROMIS® scores would discriminate between children with arthrogryposis and a pediatric reference population, and that children with arthrogryposis would report impaired upper extremity function and mobility, but normal pain interference with activities and normal peer relationships.

Methods

This is a retrospective cohort study of children with arthrogryposis aged 5-17 years who responded to four pediatric PROMIS® physical and psychosocial domain questionnaires (Mobility, Upper Extremity (UE) Function, Pain Interference, and Peer Relationships) during clinic visits at Shriners Hospital for Children, Northern California from April 2017-May 2019. Responses were converted to a T-score for comparison to the reference population (mean score = 50, standard deviation = 10).

Results

PROMIS® questionnaires were administered to 65 children with arthrogryposis with a mean age of 9.5 years. Participants reported moderately impaired Mobility (38±9) and UE Function (32±12), both significantly lower than the reference population; 63% reported moderate or severe mobility impairment, and 77% reported moderate or severe impairment with UE Function.

Regarding psychosocial function, participants reported excellent Peer Relationships (average 54±8), with 98% reporting excellent or good Peer Relationships. They also reported normal Pain Interference (average 49±10), with 82% reporting normal or mild Pain Interference.

Conclusions

PROMIS® effectively discriminates between children with arthrogryposis and the reference population. Children with arthrogryposis report moderately impaired physical function (Mobility and UE Function) but normal psychosocial function (Peer Relationships and Pain Interference.) PROMIS® is a useful tool to evaluate and understand the challenges that children with arthrogryposis face.

Background
Arthrogryposis is a term used to describe two or more congenital joint contractures in multiple body areas. Historically, the term was a disease diagnosis, but now it is purely descriptive as it occurs in over 200 different syndromes. Fetal akinesia (decreased fetal movement) and subsequent joint contractures are common to all types. Arthrogryposis can result from abnormalities in the peripheral or central nervous system, muscles, and connective tissue. Other factors believed to play a role in pathogenesis include reduced space in utero (e.g. multiple gestations or structural abnormalities), intrauterine vascular compromise, maternal disease, and external factors including toxins, drugs, or medications. The incidence of arthrogryposis varies widely, most likely because it is a descriptive term encompassing many syndromes, but it occurs in approximately 1:3,000 to 1:5000 live births. The two most common types of arthrogryposis are amyoplasia (also called arthrogryposis multiplex congenita, or AMC) and distal arthrogryposis (DA). Children with AMC typically have symmetric involvement of upper and lower limbs with shoulder internal rotation, elbow extension, wrist flexion, stiff fingers, and thumb in palm, along with hip dislocation, knee extension and ankle and foot equinovarus; occurrence is sporadic. DA describes a group of inheritable conditions (usually autosomal dominant); children with DA have a consistent pattern of hand and foot involvement with limited involvement of proximal joints.

Children with AMC or DA have a spectrum of impaired motion and strength due to contractures, and develop compensatory movements to achieve a higher level of physical function than may be anticipated. Those with more severe impairment are more likely to undergo orthopaedic surgical interventions. Impairment measures (such as range of motion and strength) provide information about physical function, but they do not necessarily characterize their ability to perform activities, or their psychosocial function including their ability to participate in life roles. Understanding a patient's perspective regarding their overall health, including both physical and psychosocial components, is an important component of caring for children with arthrogryposis, including the capacity to measure changes in abilities over time, or those associated with interventions. While a handful of tools have been developed to characterize the subjective experience of pediatric patients, one of the newest, most comprehensive, and most accessible is the Patient Reported Outcome Measurement Information System (PROMIS®). This tool was developed by the National Institute of Health, and its conceptual framework is rooted in the World Health Organization (WHO) tripartite model of health: physical, mental, and social health. PROMIS® evaluates the domains of body structure and function, activities, and participation as described in the World Health Organization (WHO) International Classification of Function – Children and Youth (ICF-CY). This conceptual framework has been mapped to the WHO International Classification of Functioning, Disability, and Health. This mapping and standardization of health terminology allows for comparison across multiple health measurement systems. PROMIS® has been validated and used in a variety of diseases to assess the effects of chronic medical conditions and help guide clinical decision-making. It features parent proxy versions (for children age 5–7) and self-report versions (age 8–17). The PROMIS® Short Form is easy to administer, and low burden for the respondent. Each PROMIS® Short Form includes 5–8 questions that measure a single domain of health, such as function (Upper Extremity Function and Mobility), pain (Pain Interference), and psychosocial (Peer Relationships).
A previous study of PROMIS® in a small sample of children with upper extremity arthrogryposis suggested that they had lower physical function scores than the general population, and normal scores in psychosocial domains.\(^\text{18}\) This small sample of children with heterogeneous presentations suggests a need for further investigation with a larger sample size to further substantiate the use of PROMIS® as a tool and more accurately describe patient reported function in children with arthrogryposis. The purpose of this study is to examine whether PROMIS® scores discriminate between children with arthrogryposis and the general reference population with respect to physical and psychosocial function. We hypothesized that children with arthrogryposis would report impaired upper extremity function and mobility, and normal pain interference and peer relationships compared to the reference population.

**Methods**

This is a retrospective cohort study of 65 children with arthrogryposis between the ages of 5–17 years who were evaluated at Shriners Hospital for Children, Northern California from April 2017 to May 2019. Children in the study had a diagnosis of AMC (57 children) or DA (8 children).

Demographic data (age, gender, race, and ethnicity) and information regarding surgery was obtained from the medical record to characterize the participants.

All participants completed the PROMIS® self-reported or parent-proxy questionnaires in English or Spanish during their clinic visit using computerized touch screen format. The four domains of PROMIS® assessed for both self-reported and parent-proxy were Mobility, Upper Extremity (UE) Function, Pain Interference, and Peer Relationships\(^\text{19}\). Children ages 8–17 years old completed the PROMIS® Pediatric Self-Assessment short forms (SF), while parents of children ages 5–7 years old completed the Parent-Proxy Assessment. PROMIS® Parent-Proxy questions ask the same content of the self-reported questionnaire, but the wording is changed from “me” to “my child”. The parent-proxy assessments have been validated and shown to correlate with the self-reported assessments\(^\text{17,20}\). Each domain consists of 8 questions for a total of 32 questions. Responses are converted to a T-score for comparison to a reference pediatric population (mean score = 50, standard deviation = 10). For Mobility, UE Function, and Peer Relationships, higher score indicates higher function, while for Pain Interference, a higher score indicates greater impairment. For Mobility and UE Function, a T-score ≥ 50 is within normal limits; 40–49 indicates mild impairment; 30–39 indicates moderate impairment, and 0–29 indicates severe impairment. For Peer Relationships, a T-score ≥ 50 is excellent; 40–49 is good; 30–39 is fair, and 0–29 is poor. For Pain Interference, a T-score ≤ 49 is within normal limits; 50–59 indicates mild impairment; 60–69 indicates moderate impairment, and 70–78 indicates severe impairment.

Basic statistics (mean, standard deviation) were completed for participant demographics and PROMIS® scores. Student’s T-test was used to compare mean PROMIS® scores in children with arthrogryposis and the reference population. The tests were 2 sided and all \(P\) values < 0.05 were considered significant.

**Results**

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PROMIS® questionnaires were administered to 65 children with arthrogryposis (33 boys and 32 girls) with a mean age of 9.5 ± 3.7 years; additional demographic data is included in Table 1. Fifty seven children had a diagnosis of AMC and 8 had a diagnosis of DA. 60 of the 65 participants had surgical intervention, and 59 of these had orthopaedic surgery: 26 had upper extremity surgery and 44 had lower extremity surgery; 15 had both UE and lower extremity (LE) surgery; and 6 had spine surgery (See Table 2). Common upper extremity interventions included posterior elbow capsulotomy with triceps lengthening, and dorsal carpal wedge osteotomy with extensor carpi ulnaris (ECU) to extensor carpi radialis brevis (ECRB) transfer. Common lower extremity interventions included adductor lengthening and percutaneous Achilles tendon lengthening.
Table 1
Characteristics of the Study Cohort

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Number (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>All</td>
<td>65 (100%)</td>
</tr>
<tr>
<td>Age in years (mean ± SD)</td>
<td>9.5 ± 3.7</td>
</tr>
<tr>
<td>Gender</td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>33 (51%)</td>
</tr>
<tr>
<td>Female</td>
<td>32 (49%)</td>
</tr>
<tr>
<td>Ethnicity</td>
<td></td>
</tr>
<tr>
<td>Not Hispanic/Latino</td>
<td>39 (60.0%)</td>
</tr>
<tr>
<td>Hispanic/Latino</td>
<td>25 (38.5%)</td>
</tr>
<tr>
<td>Other</td>
<td>1 (1.5%)</td>
</tr>
<tr>
<td>Race</td>
<td></td>
</tr>
<tr>
<td>White</td>
<td>8 (12.3%)</td>
</tr>
<tr>
<td>Asian</td>
<td>2 (3.1%)</td>
</tr>
<tr>
<td>Black or African-American</td>
<td>2 (3.1%)</td>
</tr>
<tr>
<td>American Indian or Alaskan Native</td>
<td>2 (3.1%)</td>
</tr>
<tr>
<td>Native Hawaiian/Other Pacific Islander</td>
<td>12 (18.5%)</td>
</tr>
<tr>
<td>Other</td>
<td></td>
</tr>
<tr>
<td>Diagnosis</td>
<td></td>
</tr>
<tr>
<td>Arthrogryposis multiplex congenita</td>
<td>57 (87.7%)</td>
</tr>
<tr>
<td>Distal arthrogryposis</td>
<td>8 (12.3%)</td>
</tr>
<tr>
<td>Surgical Intervention</td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>60 (92.3%)</td>
</tr>
<tr>
<td>No</td>
<td>5 (7.7%)</td>
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</table>
Table 2
Characteristics of Surgical Intervention

<table>
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<tr>
<th>Description</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total number of participants who underwent surgical intervention</td>
<td>60</td>
</tr>
<tr>
<td>Upper extremity intervention</td>
<td>26</td>
</tr>
<tr>
<td>Lower extremity intervention</td>
<td>42</td>
</tr>
<tr>
<td>Both upper and lower extremity intervention</td>
<td>15</td>
</tr>
<tr>
<td>Other combinations of interventions:</td>
<td></td>
</tr>
<tr>
<td>Lower extremity and spine intervention</td>
<td>3</td>
</tr>
<tr>
<td>Spine intervention alone</td>
<td>1</td>
</tr>
<tr>
<td>Spine and otolaryngology intervention</td>
<td>1</td>
</tr>
<tr>
<td>Cardiac intervention</td>
<td>2</td>
</tr>
</tbody>
</table>

Children with arthrogryposis reported moderately impaired Mobility (38 ± 9) and UE Function (32 ± 12), which were both significantly different than the reference population (p < 0.0001). 37% reported normal or mild Mobility impairment; 63% reported moderate or severe impairment. Only 23% reported normal or mild UE Function, while 77% rated their impairment as moderate or severe (Figs. 1 & 2).

Participants also reported excellent Peer Relationships, scoring within normal range (54 ± 8). 98% of participants demonstrated excellent or good Peer Relationships; only 2% reported them as fair. None reported them as poor.

Participants also reported normal Pain Interference (49 ± 10). 82% reported normal or mild Pain Interference, and 18% reported moderate or severe impairment.

**Discussion**

Children with arthrogryposis have impaired motion and strength secondary to joint contractures; however, these patients often achieve a higher level of physical function completing activities of daily living than may be anticipated\(^6\),\(^7\). As a result, impairment measures regarding physical function such as strength and range of motion may not accurately characterize the subjective experience of individuals. Understanding a patient’s perspective regarding their overall health, including both physical and psychosocial components, is an important component of caring for children with arthrogryposis. We found that PROMIS® function questionnaires discerned significant differences between children with arthrogryposis and a reference population, but psychosocial (peer relationships and pain) questionnaires did not.
A handful of tools exist that have been used to characterize the subjective experience of pediatric patients, including the Pediatric and Adolescent Outcome Data Collection Instrument (PODCI)\(^{21}\), the Pediatric Qualify of Life Scale (PedsQL)\(^{22}\), and most recently the Patient Reported Outcome Measure Information System (PROMIS®)\(^{8,9}\).

PROMIS® encompasses the WHO ICF-CY domains for people with chronic health conditions and its content has been mapped to the ICF classification system, which better allows for comparisons to be made across multiple health measurement systems. It also has been validated in a multitude of chronic conditions (both orthopaedic and non-orthopaedic), which allows for broader comparisons to be made. PROMIS® is also a low burden tool and easy to administer, as it has Short Forms for both child and parent-proxy reported questionnaires. Additionally, it can be uploaded into the electronic medical record and physicians can view the results before seeing the patient.

Using PROMIS®, we found that children and parent-proxies reported moderately impaired upper extremity function and mobility, but normal pain interference and excellent peer relationships in comparison to a reference population. Sixty three percent of children with arthrogryposis reported moderate to severe mobility impairment and 77% reported moderate to severe UE impairment. These findings are similar to a recent study of 29 children with UE arthrogryposis by Wall et al; their mean PROMIS® UE function score was 29 (31 for children with distal arthrogryposis and 22 for children with amyoplasia\(^{18}\)).

While children with arthrogryposis reported moderately impaired mobility and UE function, 82% reported normal or mild pain interference and 98% reported excellent or good peer relationships. This is also consistent with the study by Wall et al, which reported scores within normal limits\(^{18}\). These findings are also similar to previous studies exploring psychosocial health in congenital abnormalities in general. In examining the CoULD registry, Bae and colleagues found that children with UE congenital abnormalities had better peer relationships and positive emotional states compared to the general population\(^{23}\). While long-term data is lacking, there is evidence to suggest that peer relationships and pain remain similar to the general population into adulthood. In a survey of adults with arthrogryposis, Nouraei and colleagues found that self-reported quality of life for individuals with arthrogryposis was comparable to the US general population for several areas of health including emotional, pain, and social well-being\(^{24}\).

This study is retrospective in design, which facilitates the ease of data collection, but it is a limitation because some information may be missing from the patient’s medical record. We are thus limited in data analysis by what was collected at the clinical visit. Similarly, a second limitation of this study is that the data only characterizes one specific point in time. Further studies are needed to examine if and how patients’ PROMIS® scores change over time, and in response to interventions. Additionally, PROMIS® can only be used to assess patients who are 5 years old or older. A similar tool is needed to assess preschoolers. Finally, this study may have some selection bias in that children with more severe arthrogryposis are more likely to be followed clinically than those with lesser impairment.
Despite these limitations, however, this study is the largest cohort yet to examine PROMIS® scores in children with arthrogryposis, with a broad distribution of patient characteristics, including age, gender, and race, which is most likely representative of patients seen at clinical centers treating children with arthrogryposis. Our study indicates that PROMIS® is a useful tool to evaluate and understand the challenges that children with arthrogryposis face. Future applications of PROMIS® in children with arthrogryposis include correlating PROMIS® scores with objective physical exam measures and subgroup analysis of patient characteristics/demographics and PROMIS® scores. Additionally, longitudinal studies that follow PROMIS® scores over time will be useful in determining the effects of clinical and/or surgical interventions throughout childhood.

**Conclusions**

PROMIS® effectively discriminates between children with arthrogryposis and the reference population. Children with arthrogryposis report moderately impaired physical function (Mobility and UE Function) but normal psychosocial function (Peer Relationships and Pain Interference.) PROMIS® is a useful tool to evaluate and understand the challenges that children with arthrogryposis face.

**Abbreviations**

AMC: arthrogryposis multiplex congenita  
DA: distal arthrogryposis  
ECRB: extensor carpi radialis brevis  
ECU: extensor carpi ulnaris  
ICF-CY: International Classification of Functioning – Children & Youth  
LE: lower extremity  
PedsQL: Pediatric Quality of Life Scale  
PODCI: Pediatric Orthopaedic Data Collection Instrument  
PROMIS®: Patient Reported Outcomes Measurement Information System  
UE: upper extremity  
WHO: World Health Organization

**Declarations**
Ethics approval and consent to participate: This study (NCA1912R) was designated as exempt by Western IRB (the Institutional Review Board for Shriners Hospitals) because it was limited to review of information collected for purposes other than the proposed research.

Consent for publication: This study does not include any individual person’s data.

Availability of data and materials: Deidentified datasets analyzed for the purpose of the current study are available from the corresponding author on reasonable request.

Competing interests: The authors declare that they have no competing interests.

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Authors’ contributions: KM compiled and analyzed the data and contributed to writing the manuscript; MCM analyzed and interpreted the data and contributed to writing the manuscript; MAJ conceived of the study, supervised KM, and was a major contributor to writing the manuscript. All authors read and approved the final manuscript.

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References


19. PROMIS® ® Parent Proxy Item Bank v1.0 - Physical Function Mobility Short Form 8, PROMIS® ® Parent Proxy Item Bank v1.0 - Pain Interference Short Form 8, PROMIS® ® Parent Proxy Item Bank v1.0 - Peer Relationships Short Form 7, PROMIS® ® Parent Proxy Item Bank v1.0 - Physical Function Upper Extremity Short Form 8.


**Figures**

**Figure 1**

PROMIS® Subdomain Scores.

On average, subjects reported moderate impairment with Mobility and UE Function, and normal Peer Relationships and Pain Interference.
Figure 2

Distribution of PROMIS® Subdomain Scores.

The majority of subjects reported moderate or severe impairment with Mobility and UE Function, and normal or mild impairment with Peer Relationships and Pain Interference.