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Qualitative research in children and parents of children with achondroplasia to cognitively debrief three patient-reported outcome measures and confirm the content validity of a clinically assessed measure

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Introduction
Achondroplasia (ACH) is the most common form of short-limbed dwarfism affecting between 1 in 15 000 to 1 in 30 000 live births.1,2

Participants included patient advocates and patient advocacy groups and were assessed and consented by Health Outcomes Solutions (HOS).

Parents completed a background questionnaire containing demographic and clinical questions.

Combined concept elicitation and cognitive debriefing interviews were conducted by HOS via Zoom using semi-structured interview guides developed specifically for this study (one for children, one for adolescents, and one for parents), with feedback from advocacy groups and key opinion leaders.

In the concept elicitation portion of the interview:

– Parents were asked what medical/physical challenges their child experienced as a result of having ACH and how their child’s life was impacted by having ACH;
– Children/adolescents with ACH were asked about the medical challenges they faced and how their life was impacted as a result of having ACH.

Data from the interviews were used to map the concepts of importance to the content of the Pediatric Quality of Life Inventory® (PedsQL®),3 the Quality of Life of Short Stature Youth (QoLISSY) questionnaires,4 a pain numeric rating scale (Pain-NRS), and the Functional Independence Measure for Children (WeeFIM).5

Participants completed and provided feedback on the PedsQL, QoLISSY, and Pain-NRS measures. This did not include the clinician-administered WeeFIM; however, concepts mentioned by participants were mapped to content of the WeeFIM.

All interviews were recorded and transcribed for analysis purposes. Data from all interviews were coded using MAXQDA, a qualitative data analysis software. Coding dictionaries were developed based on the age of the child and used in the analysis of the transcripts.

The study was approved by a central institutional review board, the WCG IRB, and all interviews were recorded and transcribed for analysis purposes. Data from all interviews were coded using MAXQDA, a qualitative data analysis software. Coding dictionaries were developed based on the age of the child and used in the analysis of the transcripts.

Methods

In the concept elicitation portion of the interview:

– Conduct gap analysis to evaluate whether most concepts/symptoms are covered by these measures.
– Map concepts identified in the concept elicitation portion of the interviews to available questionnaires in future studies.

Here we report the results of the cognitive debriefing portion of the interviews. The aims were to:

– Validate the interview tool (PedsQL and QoLISSY) used in future studies.
– Understand the constructs being measured by the questionnaires.
– Identify improvements to the existing questionnaires.

Participants were found the PedsQL easier to understand than the QoLISSY (Figure 2); in general, all three patient-reported measures were found to be content valid, clear, comprehensive, and relevant for use in studies with children with ACH. The WeeFIM covers important functional concepts not covered by other patient-reported outcome measures.

Box 1. Pediatric Quality of Life Inventory® (PedsQL®)

– The PedsQL is a standardized, age-appropriate measure of health-related quality of life that captures the impact of a child’s health condition on the child’s daily life.
– The PedsQL is available in 30 languages and is used in over 40 countries.
– The PedsQL is one of the most widely used pediatric health outcomes measures.

Box 2. Quality of Life of Short Stature Youth (QoLISSY)

– The QoLISSY is a self-report measure designed to assess the quality of life of children and adolescents with short stature.
– The QoLISSY is available in 10 languages and is used in over 10 countries.
– The QoLISSY is one of the most widely used measures of quality of life for children with short stature.

Box 3. Functional Independence Measure for Children (WeeFIM)

– The WeeFIM is a clinician-administered measure of functional independence that assesses the child’s ability to perform daily activities.
– The WeeFIM is available in 12 languages and is used in over 12 countries.
– The WeeFIM is one of the most widely used measures of functional independence for children.

Results

Demographic and clinical characteristics

Eight children/adolescents were interviewed. All were Caucasian, half were female, and their mean age was 15 years (Table 1). All had undergone a surgery/procedure for ACH; four had undergone limb-lengthening surgery.

Eighteen parents of children with ACH were interviewed. Most were Caucasian, married, and had a college degree or higher (Table 2).

Table 1. Demographic and clinical characteristics at enrollment: Children/adolescents

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Children/adolescents (n=8)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex, n (%)</td>
<td>Female (5), Male (3)</td>
</tr>
<tr>
<td>Mean age, years (SD) [range]</td>
<td>13.3 (3.1) [11–16]</td>
</tr>
<tr>
<td>Race, n (%)</td>
<td>Caucasian (8)</td>
</tr>
<tr>
<td>Most common comorbid conditions, n (%)</td>
<td>Pain (8), Back pain (5), Weakness (4), Obesity (3), Sleep apnea (1)</td>
</tr>
<tr>
<td>Type of surgery, n (%)</td>
<td>None (3), Spinal stenosis (3), Pressure-skin equalization tube placement (3)</td>
</tr>
<tr>
<td>Most common medication, n (%)</td>
<td>Pain (6), Gabapentin (4)</td>
</tr>
</tbody>
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Table 2. Demographic characteristics at enrollment: Parents

<table>
<thead>
<tr>
<th>Characteristic</th>
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<tbody>
<tr>
<td>Sex, n (%)</td>
<td>Female (10), Male (6)</td>
</tr>
<tr>
<td>Mean age, years (SD) [range]</td>
<td>42.2 (6.4) [33–58]</td>
</tr>
<tr>
<td>Race, n (%)</td>
<td>Caucasian (14), Asian (1)</td>
</tr>
<tr>
<td>Work status, n (%)</td>
<td>Work full time (9), Not working for pay (7)</td>
</tr>
<tr>
<td>Mean no. of children (SD) [range]</td>
<td>2.7 (1.4) [1–6]</td>
</tr>
<tr>
<td>Mean no. of children with ACH (SD)</td>
<td>1.8 (1.0) [0–4]</td>
</tr>
</tbody>
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PedQL and QoLISSY

It is general, participants found the PedsQL and QoLISSY to be easy to complete, to contain appropriate recall period (past 24 hours) and formatting (Figure 3).

Pain-NRS

On a scale ranging from 0 to 10, where 0 represents no pain and 10 represents the worst pain possible, the mean score was 1.65 (SD 1.16) for past 24 hours as reported by both parents and children/adolescents. Pain was reported as ever having been experienced by 85% of parents and 88% of children/adolescents.

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Figure 1. Cognitive debriefing findings: PedsQL and QoLISSY

Figure 2. Comparison of PedsQL and QoLISSY

Figure 3. Cognitive debriefing findings: Pain-NRS

Mapping to questionnaires

Results of the mapping exercise indicated that the QoLISSY, PedsQL, and Pain-NRS cover most of the important concepts mentioned by parents and children/adolescents in the concept elicitation portion of the interviews (Poster #PSAT103).

Although some items from the WeeFIM were not relevant to individuals with ACH, the item relating to bladder/bowel management and items assessing mobility in terms of using the toilet and getting in and out of the bathtub, the WeeFIM covers many important concepts, such as difficulty bathing, dressing, and toiletting, not covered in other questionnaires. Therefore, it is still recommended as a clinician-reported measure in future studies.

QoLISSY, PedsQL, Pain-NRS, and WeeFIM can be used with confidence in QED-sponsored studies in ACH. Consideration should be given to using a 7-day recall period for the Pain-NRS, as pain may not be a daily occurrence.

Conclusions

This qualitative research with children and adolescents with ACH and parents of children with ACH confirmed the content validity of the PedsQL, QoLISSY, Pain-NRS, and WeeFIM.

Based on the mapping exercises, the clinician-completed WeeFIM covers many issues that are important to children and adolescents with ACH and their parents.

In general, all three patient-reported measures were found to be content valid, clear, comprehensive, and relevant for use in studies with children with ACH. The WeeFIM covers important functional concepts not covered by other patient-reported outcome measures.

The psychometric properties of these measures should be evaluated in a population with ACH.

Potential limitations of this research are that only one population subgroup (mostly Caucasian) is represented, and interviews were conducted in the US only.

Acknowledgements

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References


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