Qualitative research and parent-centric research in children and parent-centric research in children with achondroplasia: Medical challenges and their assessment using patient-reported outcome measures

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Children/adolescents

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Table 2. Demographic characteristics – parents (n=18)

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>No. of participants (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex</td>
<td>16 (89)</td>
</tr>
<tr>
<td>Mean age, years (SD) [range]</td>
<td>42.2 (6.4) [33–58]</td>
</tr>
<tr>
<td>Race</td>
<td>100</td>
</tr>
<tr>
<td>Work status</td>
<td>85 (46)</td>
</tr>
<tr>
<td>Mean no. of children (SD) [range]</td>
<td>2.7 (4.4) [0–10]</td>
</tr>
</tbody>
</table>

Cognitive debriefing results

PedQL and QoLISSY were found to be content valid, comprehensive, and reliable.

- 67% of participants found the Pain-NRS, easier to understand than the QoLISSY. However, 61% of participants found the QoLISSY to be more relevant.
- Roughly equal numbers of participants would recommend the QoLISSY and the PedQL if given a choice (42% and 46%, respectively), and 79% of participants would include both questionnaires in ACH studies (see Figures 3 and 4).
- 83% of parents reported that their child experienced pain at some time and 88% of children/adolescents reported experiencing pain at some time.
- All participants were able to accurately paraphrase the Pain-NRS and its response options. In general, participants would recommend both questionnaires in ACH studies (see Figures 3 and 4).

Figure 1. Most common medical/physical challenges reported

Figure 2. Most common impacts reported

Figure 3. Cognitive debriefing findings – PedQL, QoLISSY, and Pain-NRS

Figure 4. Comparison of PedQL versus QoLISSY

Mapping to questionnaires

- Results indicate that the QoLISSY, PedQL, Pain-NRS, and WeeFIM cover most of the important concepts:
  - >20% of parents (ranges: 21–100%) and >25% of children (ranges: 25–75%) responded with at least “moderately/ quite often” on most QoLISSY items.
  - >20% of parents (ranges: 22–78%) and >25% of children (ranges: 25–48%) responded with at least “sometimes” on most PedQL items.
  - Participants reported >30% of participants (38% of children and 28% of adults reported a score of ≥3 on the Pain-NRS.
  - WeeFIM covers important concepts such as difficulty breathing reported by 58% (38% of parents), uncoordinated gait (84% of parents, 56% of children), difficulty swallowing (50%) and difficulty remembering/remembering (50%), and lack of physical activity.

Conclusions

Data from children/adolescents and ACH and parents of children with ACH confirm the content validity of the PedQL, QoLISSY, Pain-NRS, and WeeFIM measures.

In general, the PedQL, QoLISSY, and Pain-NRS were found to be clear, relevant, and valid for use in studies of children with ACH. The WeeFIM covers important functional concepts not covered by the three PRO measures.

Potential limitations of this research are that mostly Caucasians were represented, and interviews were conducted in the US only.

References

2. 83% of parents reported that their child experienced pain at some time and 88% of children/adolescents reported experiencing pain at some time.
3. WeeFIM covers important concepts such as difficulty breathing reported by 58% (38% of parents), uncoordinated gait (84% of parents, 56% of children), difficulty swallowing (50%) and difficulty remembering/remembering (50%), and lack of physical activity.

Concept elicitation results

- Parents were asked what medical or physical challenges their child experienced as a result of having ACH. Children’s/ adolescents’ responses were asked what medical or physical challenges they experience:
  - Parents and children reported similar medical/physical challenges with some exceptions. For example, 50% of parents reported their child had sleep apnea, whereas only 17% of children reported this (see Table 2).
  - In general, (8–12 years of age) and adolescents (13–17 years of age) reported similar symptoms with some exceptions. More children reported problems with bathing (70% vs 0%), walking (100% vs. 50%), and dressing (50% vs 0%) compared with adolescents.

Acknowledgements

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Demographic and clinical characteristics

Eighteen parents of children with ACH were interviewed. Most were female, Caucasian, married, and had a college degree. On average, and one child with ACH (see Table 2).

Table 1. Demographic characteristics – children/adolescents (n=8)

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>No. of participants (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex</td>
<td>100</td>
</tr>
<tr>
<td>Mean age, years (SD) [range]</td>
<td>11.6 (1.7) [8–13]</td>
</tr>
<tr>
<td>Race</td>
<td>100</td>
</tr>
<tr>
<td>Work status</td>
<td>85 (46)</td>
</tr>
<tr>
<td>Mean no. of children (SD) [range]</td>
<td>1.5 (1.0) [0–3]</td>
</tr>
</tbody>
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Intervention

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

- Individuals with ACH experience various medical and physical symptoms and impacts, which may affect their quality of life.
- As part of the qualitative research study, combined concept elicitation (CE) and cognitive debriefing (CD) interviews were conducted in children/adolescents with children with ACH:
  - Better understand the symptoms and impacts experienced by individuals with ACH.
  - Evaluate the clarity, relevance, and appropriateness of three patient-reported outcome (PRO) measures and one parent-reported outcome measure for use in ACH studies.

Methods

- Participants in the US were identified by patient advocates and patient advocacy groups.
- Health Outcomes Solutions (HOS) screened and obtained informed consent from all participants.
- Children/adolescents who took part in the study were 8–17 years of age, with a confirmed molecular diagnosis of ACH. Parents were required to have a child 2–17 years of age with a confirmed molecular diagnosis of ACH.
- Parents completed a background questionnaire containing demographic and clinical questions.
- Following interviews were conducted by HOS via Zoom using a semi-structured interview guide developed specifically for the study (one for children, one for adolescents, and one for parents):
  - During the CE portion of the interview, participants were asked about medical and physical challenges experienced and how those challenges impacted their life or their child’s life.
  - During the CD portion of the interview, participants were asked to complete and provide feedback on the Pediatric Quality of Life Inventory (PedsQL), the Quality of Life of Short Stature Youth (QoLISSY), and the Pain-Numeric Rating Scale (NRS).
- Advocates, advocacy groups, and key opinion leaders provided input on the study protocol and interview guides.

Concepts identified during the CE portion of the interview were mapped to the PedsQL, QoLISSY, Pain-NRS, and a clinician-administered measure, the Functional Independence Measure for Children (WeeFIM).

The following proportions were calculated for each item to determine coverage:
- Percent scoring at least “sometimes” on the PedsQL, (response never, almost never, somewhat, often, almost always).
- Percent scoring at least “moderately” on the QoLISSY: responses not at all, slightly/waistly, moderately/quite often, very/very often, and extremely/extremely.
- Percent reporting ≥30 on Pain-NRS responses: (0 no pain) to 10 (worst pain possible).

The study was approved by a central institutional review board, the WCG IRB.