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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#PSAT102

Introduction

- 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.[1,2]

- Individuals with ACH experience various medical symptoms/complications and impacts during their lifetime.

- We conducted combined concept elicitation and cognitive debriefing interviews in children with ACH and parents of children with ACH to better understand ACH-related symptoms/complications and their impacts on health-related quality of life in this population.

- Here we report the results of the cognitive debriefing portion of the interviews. The aims were to:
  - Map concepts identified in the concept elicitation portion of the interviews to available patient-reported outcomes measures.
  - Conduct gap analysis to evaluate whether most concepts/symptoms are covered by these measures.

- The results of the concept elicitation interviews are presented in poster #PSAT103.

Methods

- Participants were identified by patient advocates and patient advocacy groups and were screened 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) questionnaire,[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. 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 conducted in the USA.

Results

Demographic and clinical characteristics

- Eight children/adolescents were interviewed. All were Caucasian, half were female, and their mean age was 13 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 female, Caucasian, married, and had a college degree or higher (Table 2).

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

Characteristic	Children/adolescents (N=8)
Sex, n (%)	
Female	4 (50)
Mean age, years (SD) [range]	13.3 (2.0) [11–16]
Race, n (%)	
Caucasian	8 (100)
Most common comorbid conditions, n (%)	
Back pain	5 (63)
Hydrocephalus	4 (50)
Obesity	3 (38)
Spinal stenosis	3 (38)
Type of surgery, n (%)[a]	
Tonsillectomy/adenoidectomy	4 (50)
Pressurization equalization tube placement	3 (38)

[a]Most commonly reported.
SD = standard deviation.

Table 2. Demographic characteristics at enrollment: Parents

Characteristic	Parents (N=18)
Sex, n (%)	
Female	16 (89)
Mean age, years (SD) [range]	42.2 (6.4) [33–58]
Race, n (%)	
Caucasian	16.5 (92)
Asian	1.5 (8)
Work status, n (%)	
Work full time/part time	6 (33)/3 (17)
Not working for pay/other	9 (50)
Mean no. of children (SD) [range]	2.7 (6.4) [1–5]
Mean no. of children with ACH (SD)	1 (0)

SD = standard deviation.

PedsQL and QoLISSY

- In general, participants found the PedsQL and QoLISSY to be easy to complete, to contain relevant items, and to be formatted appropriately (Figure 1).

- "Past month" was an acceptable recall period for PedsQL and "past week" was appropriate for QoLISSY, although only 45% found it easy to think about the past week for QoLISSY.

- Participants found the PedsQL easier to understand than the QoLISSY (Figure 2); however, 61% found the content of QoLISSY to be more relevant to individuals with ACH. Overall, 79% would recommend including **both** questionnaires in future studies.

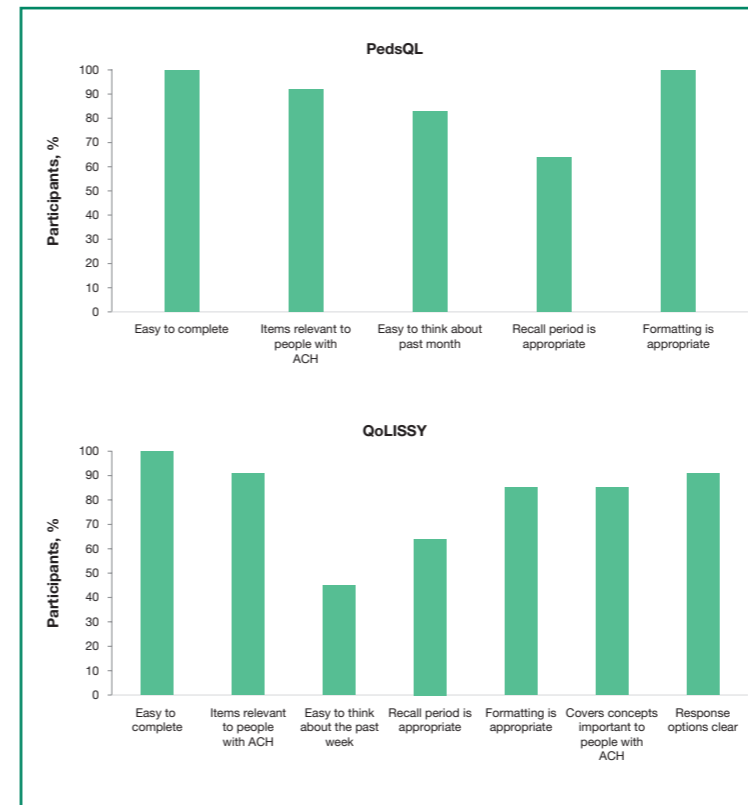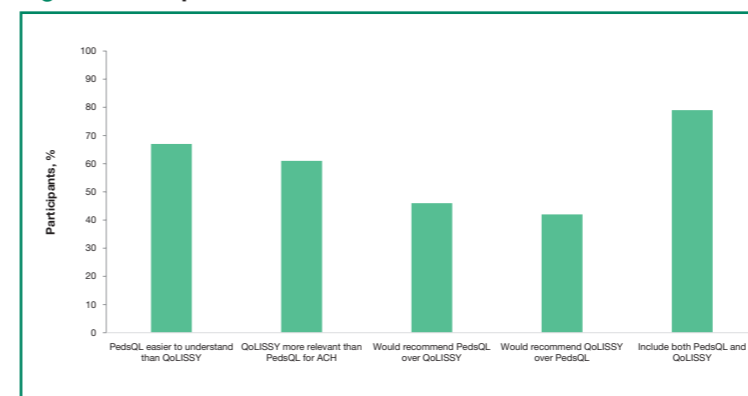**Figure 1.** Cognitive debriefing findings: PedsQL and QoLISSY

Figure 2. Comparison of PedsQL and QoLISSY

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 1.6 (SD 1.9) for past 24 hours as reported by both parents and children/adolescents. Pain was reported as ever having been experienced by 83% of parents and 88% of children/adolescents.

- Participants found the Pain-NRS to be clear, relevant, and easy to complete, with an appropriate recall period (past 24 hours) and formatting (Figure 3).

Figure 3. Cognitive debriefing findings: Pain-NRS
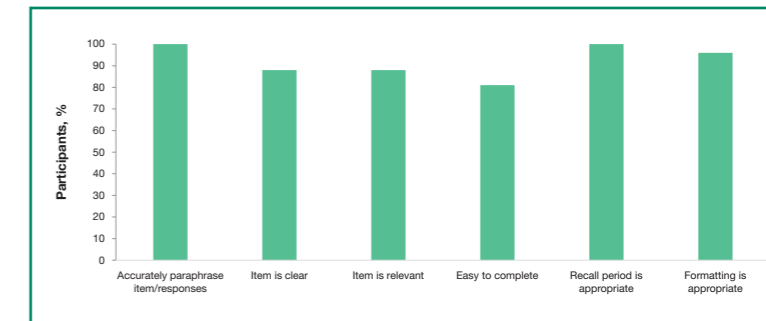
Mapping to questionnaires

- 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 (e.g., items 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 contains many important concepts, such as difficulty bathing, dressing, and toileting, 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 exercise, 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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