

A background review of several generic patient-reported outcome and clinician-reported outcome measures: Selecting measures for further evaluation in a qualitative study in achondroplasia

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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 and physical symptoms and impacts (social, emotional, etc.), which may affect their quality of life.^{3,4}
- No validated ACH-specific patient-reported outcome (PRO) or clinician-reported outcome (ClinRO) measures are currently available for use.

Objectives

- To conduct a review of the literature to determine the medical, physical, and emotional challenges and impacts experienced by children with ACH to identify potential PROs and ClinROs.
- To determine content coverage and appropriateness of potential PROs and ClinROs for use in upcoming ACH studies.

Methods

- Published literature was reviewed to identify the most important medical, social, and emotional challenges and impacts for children with ACH.
- Various generic outcomes instruments were evaluated for relevance based on these findings:
 - PRO measures: Pediatric Quality of Life Inventory (PedsQL), Quality of Life of Short Stature Youth (QoLISSY), Childhood Health Assessment Questionnaire (CHAQ), 45-item Child Health Questionnaire self-report Child Form (CHQ-CF45)/50-item Child Health Questionnaire self-report Parent Form (CHQ-PF50), Pediatric Outcomes Data Collection Instrument (PODCI), and Pain Numeric Rating Scale (Pain-NRS).
 - ClinRO measure: Functional Independence Measure for Children (WeeFIM).
- A mapping exercise was conducted using data from published studies to evaluate concept coverage for each measure. Each measure was reviewed in detail, including evaluating the availability of measures (parent vs. child report), number of items, and evidence of psychometric properties.
- Measures were then selected for further evaluation during a qualitative study (combined concept elicitation and cognitive debriefing interviews) with children/adolescents with ACH and parents of children with ACH (see ESPE 2022 poster #P1-327).

Results

Mapping of concepts to identified measures

- Analysis of available published literature showed that all identified measures (QoLISSY, PedsQL, CHAQ, CHQ-CF45/CHQ-PF50, PODCI, WeeFIM, and Pain-NRS) had good concept coverage of symptoms and impacts (physical, social, emotional, school, etc).⁵⁻¹¹
- Table 1 shows the concepts that were reported with high frequency (22–100%) in the literature by patients of children with ACH, and which measures cover each concept.

Pros and cons of reviewed measures in terms of concept coverage

- Table 2 includes a comparison of the advantages (pros)/disadvantages (cons) of each measure in terms of concept coverage.
- Although all measures contain some important concepts relevant to individuals with ACH, no single measure covers all concepts.

Table 1. Concepts commonly reported by parents of children with ACH

Concept	Covered in
Needing assistance from others outside/at school	QoLISSY, CHAQ, PODCI
Feeling sad, depressed, frustrated, angry, annoyed	QoLISSY, PedsQL, CHQ-CF45/CHQ-PF50
Feeling different	QoLISSY, PedsQL, CHQ-CF45/CHQ-PF50, PODCI
Being treated as younger than age	QoLISSY
Having to explain ACH to others	QoLISSY
Difficulty reaching things	QoLISSY, CHAQ
Difficulty running, walking, climbing stairs, participating in sports	QoLISSY, PedsQL, CHAQ, CHQ-CF45/CHQ-PF50, WeeFIM
Difficulty with tasks requiring fine motor control	QoLISSY, CHAQ, PODCI
Difficulty toileting, bathing, grooming, dressing	QoLISSY, PedsQL, CHAQ, PODCI, WeeFIM
Being teased/bullied	QoLISSY, PODCI, PedsQL, CHQ-CF45/CHQ-PF50
Pain	CHAQ, PedsQL, CHQ-CF45/CHQ-PF50, PODCI, Pain-NRS
Needing adaptive devices	PODCI, CHAQ

Table 2. Pros/cons of reviewed measures

Questionnaire	PROs	Cons
QoLISSY	<ul style="list-style-type: none"> Both child and parent versions available Developed for individuals with short stature so questions are more targeted toward individuals with ACH Comprehensive for assessing emotional/social impacts related to height 	<ul style="list-style-type: none"> Many concepts related to physical functioning, dressing/undressing, tasks requiring fine motor skills, toileting not included Does not assess impacts at school and need for adaptations or assistance
PedsQL	<ul style="list-style-type: none"> Various versions available based on age of child; parent version also available Assesses three key symptoms: pain, low stamina/tiring easily, and sleep apnea Assesses many emotional impacts, some social impacts, and impacts at school 	<ul style="list-style-type: none"> Does not assess all emotional impacts Does not assess some of the highly endorsed concepts, such as toileting and reaching high objects, and need for adaptations or assistance
CHAQ	<ul style="list-style-type: none"> Both parent and child versions available Assesses many ADLs (dressing, toileting, bathing), physical functioning, and need for assistance and use of adaptations 	<ul style="list-style-type: none"> Does not assess emotional or social impacts and impacts at school
CHQ-CF45/CHQ-PF50	<ul style="list-style-type: none"> Assesses two key symptoms: pain and sleep apnea Assesses many aspects of physical functioning, some school impacts, and many emotional and social impacts 	<ul style="list-style-type: none"> Does not assess need for assistance/use of adaptive devices
PODCI	<ul style="list-style-type: none"> Includes versions for parents and adolescents Assesses three key symptoms: pain, low energy/stamina, and sleep apnea Assesses some school impacts, many aspects of physical functioning, many social impacts, and many ADLs (dressing, bathing, toileting) 	<ul style="list-style-type: none"> Does not assess most emotional impacts or need for assistance or use of adaptive devices
WeeFIM	<ul style="list-style-type: none"> Assesses self-care activities such as bathing/grooming, and toileting Assesses some aspects of physical functioning, but not comprehensively 	<ul style="list-style-type: none"> Does not assess emotional impacts, impacts at school, need for assistance, or use of adaptive devices Is limited in assessing social impacts
Pain-NRS	<ul style="list-style-type: none"> Assesses pain 	<ul style="list-style-type: none"> Does not assess any other important concepts

ADL: activities of daily living.

Summary of measures reviewed

- A detailed review of each measure was conducted, and a recommendation was made as to whether the measure should be evaluated further in a qualitative study of children/adolescents with ACH and parents of children with ACH (see Table 3 for further details).

Table 3. Summary of measures reviewed

Measure	Assessment	No. of items	Concepts	Psychometric properties demonstrated	Recommended – Yes/No
CHAQ	Change in physical functioning in children with arthritis	30	Dressing/grooming, arising, eating, walking, hygiene, reach, grip, activities	Some	No – covers some key concepts but only a parent version available
CHQ-CF45/CHQ-PF50	Physical and psychological functioning and well-being in children/adolescents	45/50	Global health, physical functioning, social/emotional, social/physical, body pain, getting along, global behavior, self-esteem, mental and general health, change in health, family activities, family cohesion	Not reported	No – lengthy questionnaire and psychometric properties not evaluated
PedsQL	HRQoL in healthy children/adolescents and those with acute/chronic conditions	23	Physical, emotional, social, school functioning	Some	Yes – covers physical, emotional, social, and school functioning
PODCI	Changes following pediatric orthopedic interventions for range of diagnoses	83–86	Upper extremity and physical function, transfer and mobility tasks, sports/physical functioning, pain/comfort, treatment expectations, happiness, satisfaction with symptoms	Not reported	No – lengthy questionnaire and psychometric properties not evaluated
QoLISSY (Core)	HRQoL in children/adolescents with short stature	22	Physical, social, emotional functioning	Majority	Yes – covers physical, social, and emotional functioning in children with short stature
WeeFIM	Need for assistance and severity of disability in children	18	Self-care, mobility, cognition	Some	Yes – covers areas of self-care not included in other measures
Pain-NRS	Pain severity	1	Pain	Not reported	Yes – commonly used to assess pain severity

HRQoL: health-related quality of life.

Conclusions

- The QoLISSY, PedsQL, Pain-NRS, and WeeFIM were identified for further evaluation in a qualitative study of children with ACH and parents of children with ACH, based on this review of the literature.

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