

Qualitative research in children with achondroplasia and parents of children with achondroplasia: medical challenges and impacts

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#PSAT103

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 concept elicitation portion of interviews in children/adolescents with ACH and parents of children/adolescents with ACH. The results of the cognitive debriefing portion of the interviews are presented in poster #PSAT102.

Methods

- Participants were identified by patient advocates and patient advocacy groups and were screened and consented by Health Outcomes Solutions (HOS).
- Children/adolescents between 8 and 17 years of age with confirmed molecular diagnosis were eligible to participate.
- Parents completed a background questionnaire containing demographic and clinical questions.
- The percentages of medical challenges (ACH-related symptoms/complications) and impacts of these challenges to HRQoL were calculated separately for children and parents.
- 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.
- 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.
- Saturation tables were developed to categorize each challenge/impact mentioned by each participant. Saturation, the point at which no new concepts are mentioned by subsequent participants, should ideally be achieved to confirm content validity.
- 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)

^aMost 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	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.

Concept elicitation findings: Medical/physical challenges reported by children/adolescents and parents (Figure 1)

- The challenges most commonly reported by children/adolescents were feeling hot/sweaty (88%), pain (88%), balance issues (75%), fatigue (63%), muscle fatigue/loose joints (63%), and speech issues (63%). With the exception of sleep apnea, saturation was reached by the 8th interview.
- The challenges most commonly reported by parents were pain (83%), ear infections (78%), and feeling hot/sweaty (78%). With the exception of bed wetting, saturation was reached by the 18th interview.
- Some differences were observed between parents and children/adolescents in their reporting of medical challenges. Ear infections and sleep apnea were more commonly reported by parents, whereas children/adolescents more often reported muscle fatigue/loose joints and fatigue as challenges faced.

Figure 1. Most commonly reported medical challenges

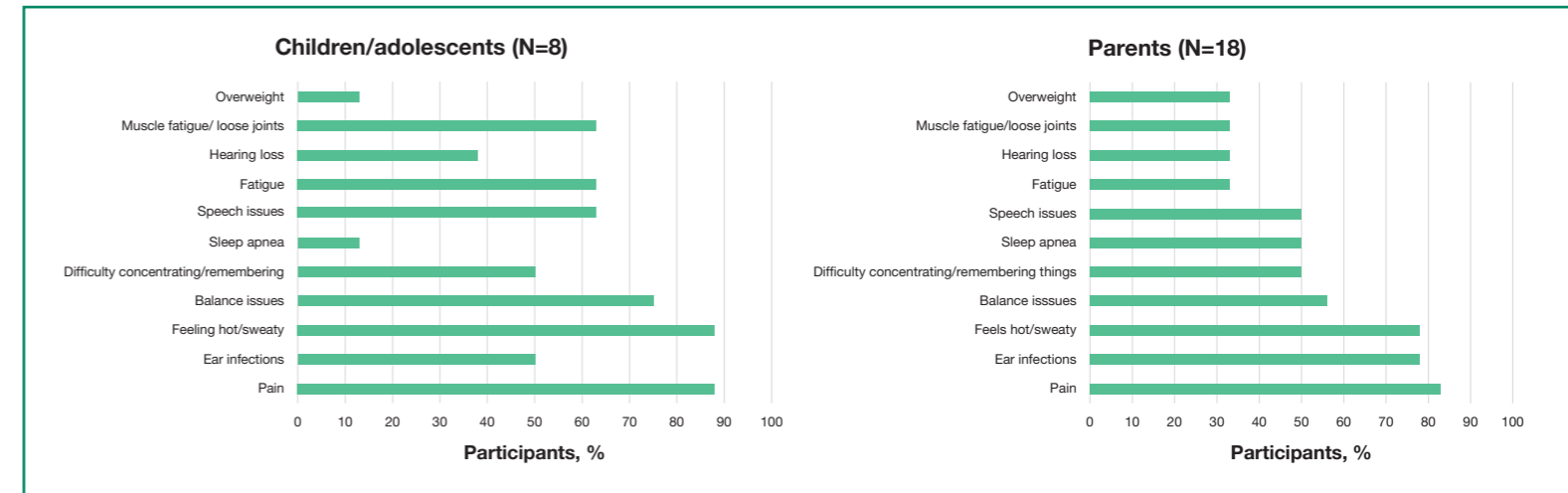
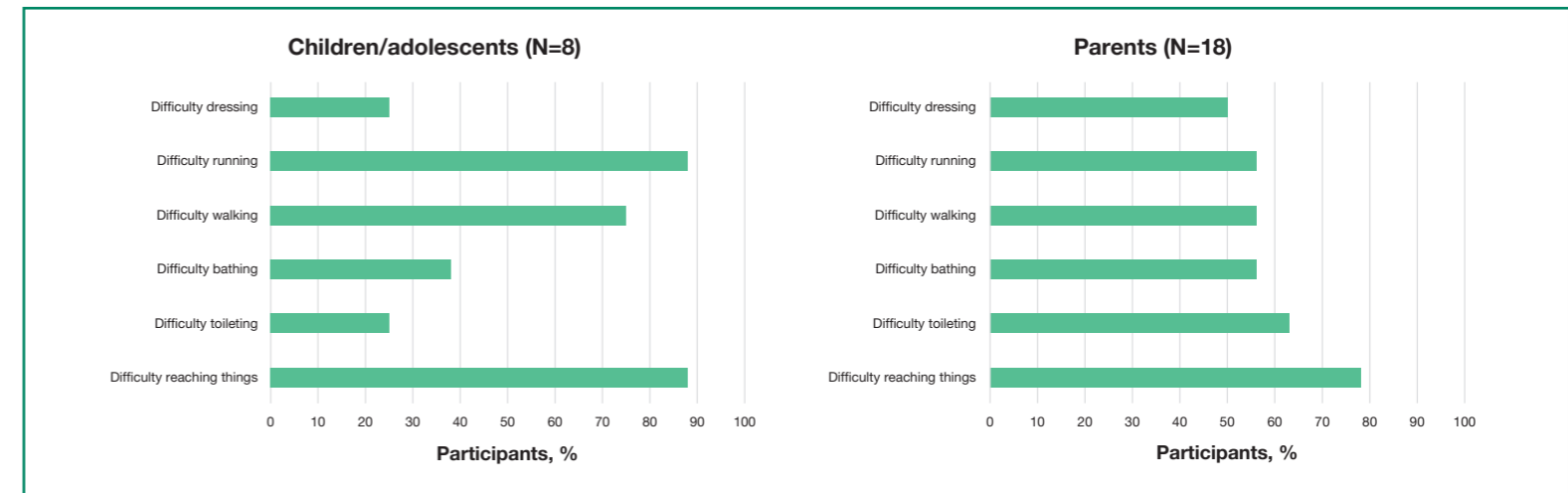


Figure 2. Most commonly reported impacts



Concept elicitation findings: Impacts reported by children/adolescents and parents (Figure 2)

- The impacts most commonly reported by children/adolescents were difficulty reaching things (88%), running (88%), and walking (75%). Saturation was reached by the 7th interview.
- The impacts most commonly reported by parents were difficulty reaching things (78%), toileting (63%), bathing (56%), walking (56%), running (56%), and dressing (50%) (Figure 2). Saturation was reached by the 9th interview.
- Some differences were observed between parents and children/adolescents in the impact of ACH. Difficulties with toileting, dressing, and bathing were more commonly reported by parents, whereas children/adolescents more often reported difficulties running and walking.

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Conclusions

- This research provides detailed information on the medical challenges and impacts faced in everyday life by children with ACH.
- Individuals affected by ACH experience numerous physical and medical challenges and impacts as a result of their condition.
- Potential limitations of this research are that only one population subgroup (mostly Caucasian) is represented, and interviews were conducted in the US only.
- The concepts identified with high frequency were mapped to patient-reported outcome and functional measures to identify the most appropriate and relevant measures to include in QED-sponsored studies of ACH. See poster no. PSAT102 for more details.

References

- Horton WA, et al. Lancet 2007;370:162–72.
- Waller DK, et al. Am J Med Genet A 2008;146A:2385–9.