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

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, Inc. (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 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 subsequent participants, should ideally be achieved to confirm content validity.

The results of the cognitive debriefing portion of the interviews are presented in poster #PSAT102.

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).

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 (38%), and speech issues (38%). With the exception of speech apnea, saturation was reached by the 9th interview.

The challenges most commonly reported by parents were pain (50%), air infections (78%), and feeling cold/wet/sweaty (78%). With the exception of bad teething, saturation was reached by the 10th 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.

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

The impacts most commonly reported by children/adolescents were difficulty reaching things (68%), running (38%), and sleeping (78%). Saturation was reached by the 7th interview.

The impacts most commonly reported by parents were difficulty reaching things (78%), toileting (50%), and bathing (50%). Saturation was reached by the 10th 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.

Acknowledgements

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Conclusions

This research provided 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 GED-sponsored studies of ACH. See poster no. PSAT102 for more details.

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

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Children/adolescents (N=8)</th>
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</thead>
<tbody>
<tr>
<td>Sex, n (%)</td>
<td>Female 4 (50)</td>
</tr>
<tr>
<td>Mean age, years (SD) [range]</td>
<td>13.3 (2.0) [11–16]</td>
</tr>
<tr>
<td>Race, n (%)</td>
<td>Caucasian 1 (10)</td>
</tr>
<tr>
<td>Most common comorbid conditions, n (%)</td>
<td>Back pain 5 (63)</td>
</tr>
<tr>
<td>Type of surgery, n (%)</td>
<td>Tonsillectomy/adenoidectomy 4 (50)</td>
</tr>
</tbody>
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Table 2. Demographic characteristics at enrollment: Parents

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Parents (N=18)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex, n (%)</td>
<td>Female 16 (89)</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 18 (94)</td>
</tr>
<tr>
<td>Work status, n (%)</td>
<td>Work full time/part time 6 (33) [17]</td>
</tr>
<tr>
<td>Mean no. of children (SD) [range]</td>
<td>2.7 (0.6) [1–5]</td>
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</table>

Figure 1. Most commonly reported medical challenges

Figure 2. Most commonly reported impacts

References