Development of a Measure for the Impacts of Pediatric Achondroplasia on Children's Daily Functioning and Well-being

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BACKGROUND

The purpose of the study was to investigate the impacts on children’s daily lives to support the development of the Achondroplasia Child Experience Measure – Impact (ACEM – Impact), which assesses the impact of the functioning and well-being of children aged 2 to 12 years.

METHODS

The qualitative research study design was based on an adapted grounded theory approach and followed FDA guidelines for the development of patient-reported outcome measures (PROMs). Based on a literature review and clinical expert interviews, a semi-structured interview guide was developed to elicit parents’ experiences related to ACH.

Objective

The qualitative analysis and the development of a preliminary theoretical model identified four conceptual domains for the impacts of ACH, as well as major impacts in each domain:

- Functioning and daily life, including school participation:
  - 13 major impacts (Figures 1 & 2)
  - Emotional well-being:
    - 6 major impacts (Figure 3)
  - Social well-being:
    - 7 major impacts (Figure 4)
  - Need for assistance/adaptive devices:
    - 5 major impacts (Figure 5)

RESULTS

Demographic/Health Characteristics for Children of Parent Participants

<table>
<thead>
<tr>
<th>Child’s age, n (%)</th>
<th>2 yrs</th>
<th>3 yrs</th>
<th>4 yrs</th>
<th>5 yrs</th>
<th>6 yrs</th>
<th>7 yrs</th>
<th>8 yrs</th>
<th>9 yrs</th>
<th>10 yrs</th>
<th>11 yrs</th>
<th>12 yrs</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number of parents</td>
<td>4</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>3</td>
<td>5</td>
<td>5</td>
<td>2</td>
<td>2</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Health status, n(%):</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Healthy</td>
<td>12 (35.3)</td>
<td>9 (42.3)</td>
<td>14 (35.9)</td>
<td>10 (45.5)</td>
<td>5 (18.2)</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Recurrent infections</td>
<td>8 (23.5)</td>
<td>6 (26.7)</td>
<td>9 (23.7)</td>
<td>7 (31.8)</td>
<td>5 (18.2)</td>
<td>2 (10.0)</td>
<td>1 (5.0)</td>
<td>0</td>
<td>0</td>
<td>1 (5.0)</td>
<td>0</td>
</tr>
<tr>
<td>Ear problems</td>
<td>10 (28.6)</td>
<td>6 (26.7)</td>
<td>10 (26.3)</td>
<td>9 (40.9)</td>
<td>8 (29.0)</td>
<td>6 (30.0)</td>
<td>5 (25.0)</td>
<td>2 (10.0)</td>
<td>1 (5.0)</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Other sleep issues</td>
<td>6 (17.1)</td>
<td>4 (18.2)</td>
<td>6 (16.2)</td>
<td>5 (22.7)</td>
<td>7 (25.0)</td>
<td>4 (20.0)</td>
<td>4 (20.0)</td>
<td>2 (10.0)</td>
<td>2 (10.0)</td>
<td>1 (5.0)</td>
<td>0</td>
</tr>
<tr>
<td>Bowing of legs</td>
<td>12 (33.8)</td>
<td>7 (33.3)</td>
<td>12 (32.4)</td>
<td>11 (50.0)</td>
<td>8 (28.6)</td>
<td>6 (30.0)</td>
<td>7 (35.0)</td>
<td>2 (10.0)</td>
<td>2 (10.0)</td>
<td>1 (5.0)</td>
<td>0</td>
</tr>
<tr>
<td>Limited physical activity</td>
<td>10 (28.6)</td>
<td>8 (36.4)</td>
<td>9 (24.3)</td>
<td>8 (36.4)</td>
<td>7 (25.0)</td>
<td>6 (30.0)</td>
<td>7 (35.0)</td>
<td>2 (10.0)</td>
<td>2 (10.0)</td>
<td>1 (5.0)</td>
<td>0</td>
</tr>
</tbody>
</table>

Figure 1. Major impacts on children’s functioning and daily life

- Reading difficulties
  - Math difficulties
  - Writing difficulties
  - Physical activity
  - Social isolation

Figure 2. Major impacts on children’s school participation among school-aged children

- Need for medical treatment
  - Need for assistance/adaptive devices
  - Need for accommodations/adaptations

CONCLUSIONS

The newly developed ACEM – Impact measure included 31 items in 4 conceptual domains:

- The ACEM – Impact was designed as an observer-reported outcome (ObsRO) measure to be completed by parents of children aged 2 to 12 years with ACH
- Based on the cognitive debriefing interviews, minor edits to the measure were made to improve understanding and readability
- The ACEM – Impact is a validation-ready parent ObsRO designed to assess the impacts of ACH on the functioning and well-being of children aged 2 to 12 years with ACH

- The study provides evidence to support the content validity for the validation-ready ACEM – Impact parent ObsRO measure to assess the impacts of ACH on daily functioning and emotional and social well-being in children aged 2 to 12 years
- A future psychometric validation study of the ACEM – Impact is needed to further assess measure validity and reliability
- As new treatments for pediatric ACH are being developed, it is critical for clinicians to understand and assess the impacts of ACH on children’s general functioning and well-being that may be improved with treatment