Pediatric Achondroplasia: Impacts on Children's Functioning and Well-being

Kathryn M. Pfeiffer1, Menyl Brod1, Dorthe Vium1, Sha Ota1, Jill Gianotti2, Jonathan A. Leff1
1The Brod Group, 2Ascendis Pharma A/S, 3Ascendis Pharma, Inc.

RESULTS

The purpose of the study was to investigate the impacts of ACH on the functioning, daily life, and well-being of children aged 2 to <12 years.

METHODS

The qualitative research study design was based on an adapted grounded theory approach. Based on a literature review and clinical expert interviews, a semi-structured interview guide was developed to elicit parents' experiences related to ACH.

Inclusion criteria:
- adult aged 18 years or older
- able to read, write, and speak English (in the United States [US]) or Spanish (in Spain)
- parent of a child (<18 years of age) diagnosed with ACH; and
- actively involved in the child's care

Exclusion criteria:
- A cognitive impairment or other medical condition, including psychiatric conditions, that would affect a participant's ability to take part in a telephone interview or focus group
- Difficulty participating in sports/physical play
- Being physically active
- Dressing/undressing self

It should be noted that this study was part of a larger study of parents of children with ACH <18 years of age, and this study focused only on parents of children aged 2 to <12 years. Individual telephone interviews and 1 parent focus group were conducted in the US and Spain with 36 parents of children aged 2 to <12 years with ACH.

Telephone interviews lasted approximately 1 hour, and the focus group lasted 2 hours. The interviews/focus group were conducted in English (US) or Spanish (Spain), transcribed verbatim, and translated to English if necessary. Interview and focus group transcripts were analyzed for content and coded by themes using a qualitative analysis software program.

RESULTS

Parent Participant Sample Description

Participant sample characteristics are shown in Table 1.

- Average age of parents was 41.5 years (SD, 6.6), range, 32-68
- Thirty-one parents were mothers (86.1%), and 5 parents were fathers (13.9%)
- Most parents were married (80.6%, n=29), 8.3% were partnered (n=3), 5.6% were divorced (n=2), and 5.6% were single (n=2)
- Seven parents (19.4%), all residing in the US, also had a diagnosis of ACH

Table 1. Parent participant demographic characteristics

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Total (n=36)</th>
<th>US (n=31)</th>
<th>Spain (n=5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age, mean (SD)</td>
<td>41.5 (6.6)</td>
<td>41.7 (6.0)</td>
<td>40.5 (7.6)</td>
</tr>
<tr>
<td>Sex (male)</td>
<td>5 (13.9%)</td>
<td>3 (9.7%)</td>
<td>2 (40.0%)</td>
</tr>
<tr>
<td>Marital status</td>
<td>28 (77.8%)</td>
<td>26 (83.9%)</td>
<td>2 (40.0%)</td>
</tr>
<tr>
<td>Mothers</td>
<td>31 (86.1%)</td>
<td>30 (96.8%)</td>
<td>1 (20.0%)</td>
</tr>
<tr>
<td>Fathers</td>
<td>5 (13.9%)</td>
<td>1 (3.2%)</td>
<td>4 (80.0%)</td>
</tr>
<tr>
<td>Race (White)</td>
<td>34 (94.4%)</td>
<td>31 (99.1%)</td>
<td>3 (60.0%)</td>
</tr>
<tr>
<td>Education (HS+)</td>
<td>2 (5.6%)</td>
<td>2 (6.5%)</td>
<td>0 (0.0%)</td>
</tr>
<tr>
<td>Employment</td>
<td>33 (91.7%)</td>
<td>30 (96.8%)</td>
<td>3 (60.0%)</td>
</tr>
<tr>
<td>Full-time work</td>
<td>9 (25.0%)</td>
<td>7 (22.6%)</td>
<td>2 (40.0%)</td>
</tr>
<tr>
<td>Parental care</td>
<td>33 (91.7%)</td>
<td>30 (96.8%)</td>
<td>3 (60.0%)</td>
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</tbody>
</table>

Impacts on Emotional Well-being

Frequently reported impacts on children's emotional well-being related to ACH included feeling:
- Different (53%, n=19)
- Frustrated (47%, n=17)
- Depressed/sad (39%, n=14)
- Angry/irritable (30%, n=12)
- Embarrassed/self-conscious (33%, n=12)

Impacts on Social Well-being

The most frequently reported impacts on children's social well-being were:
- Difficulty participating in sports/physical play (98%, n=31)
- Being treated as younger than age (83%, n=30)
- Negative attention in public, such as staring/pointing (84%, n=23)
- Experience of teasing/bullying (64%, n=23)
- Difficulty participating in social activities, such as birthday parties/playdates (64%, n=23)

Impacts on Children's Functioning and Daily Life

The most frequently reported impacts on children's functioning and daily life were difficulty/issues with:
- Reaching objects/high places (89%, n=32)
- Toileting self (67%, n=24)
- Bathing/washing/grooming (58%, n=21)
- Running (66%, n=20)
- Walking (50%, n=18)
- Being physically active (47%, n=17)
- Dressing/undressing self (47%, n=17)
- Being popular/well-known (14%, n=5)
- Connecting with the dwarfism community (8%, n=3)
- Being stigmatized by others (8%, n=3)
- Being teased/bullied (6%, n=2)
- Hair care (2%, n=1)

Impacts on Children's School Participation

Among school-aged children (aged 5 to <12 years, n=25), challenges to school participation included:
- Missed school time (76%, n=19)
- Being physically active (47%, n=17)
- Difficulty participating in class/schoolwork (40%, n=10)
- Difficulty getting from place to place at school (32%, n=8)
- Difficulty participating in sports/physical play (22%, n=6)
- Difficulty with tasks requiring fine motor skills (14%, n=4)
- Difficulty with tasks requiring fine motor skills (14%, n=4)
- Difficulty with daily living/social skills (14%, n=4)
- Being teased/bullied (8%, n=2)
- Being popular/well-known (6%, n=2)
- Connecting with the dwarfism community (4%, n=1)
- Being stigmatized by others (4%, n=1)
- Being teased/bullied (4%, n=1)
- Hair care (2%, n=1)

Study Limitations

Given the relatively small sample size, results should be interpreted with caution. Percentage differences in parents’ reported impacts may not reflect actual differences in the population.

Results may not be generalizable to other groups/populations. For example, parents and children’s experiences related to ACH may vary in different countries, cultures, and healthcare systems.

Conclusions

To our knowledge, this is the first study to investigate the broad impacts of ACH on children's functioning and general well-being.

The findings highlight some of the difficulties that children with ACH experience in their functioning and daily life, as well as challenges to school participation.

Additionally, the study suggests that children with ACH experience a range of emotional and social impacts.

Appendix A: Supplementary data

Appendix B: Supplementary data

Appendix C: Supplementary data

Appendix D: Supplementary data

Appendix E: Supplementary data

Appendix F: Supplementary data

Appendix G: Supplementary data

Appendix H: Supplementary data

Appendix I: Supplementary data

Appendix J: Supplementary data

Appendix K: Supplementary data

Appendix L: Supplementary data

Appendix M: Supplementary data

Appendix N: Supplementary data