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

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BACKGROUND

- The clinical complications and medical impacts of achondroplasia (ACH) in children and adults are well studied[1,2]
- Frequent complications of ACH in childhood include recurrent ear infections (otitis media), sleep apnea, hearing loss, teeth crowding/misalignment, and speech delay or articulation problems, while frequent complications in adulthood include chronic back and leg pain, spinal stenosis, sleep apnea, and obesity[2,3]
- Research has also shown that infants and young children with ACH experience delays in some developmental milestones, including gross motor, fine motor, communication, and feeding milestones[4,5]
- Less is known about the broader impacts of ACH on children's lives, including impacts on functioning and daily life, emotional well-being, and social well-being

OBJECTIVE

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

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 participants 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

	Spain (n=11)	US (n=25)	Total (N=36)
Age, mean(SD)	40.4(3.1)	42.0(7.6)	41.5(6.6)
(range)	(35-43)	(32-68)	(32-68)
Relationship to child, n(%)			
mother	8(72.7)	23(92.0)	31(86.1)
father	3(27.3)	2(8.0)	5(13.9)
Marital status, n(%)			
single	2(18.2)	0	2(5.6)
married	6(54.5)	23(92.0)	29(80.6)
partnered	3(27.3)	0	3(8.3)
divorced	0	2(8.0)	2(5.6)
Education, n(%)			
less than high school	2(18.2)	1(4.0)	3(8.3)
high school or equivalent	4(36.4)	2(8.0)	6(16.7)
college degree	5(45.5)	12(48.0)	17(47.2)
post-graduate school	0	10(40.0)	10(27.8)
Work status, n(%)			
full-time	6(54.5)	10(40.0)	16(44.4)
part-time	3(27.3)	3(12.0)	6(16.7)
student	0	2(8.0)	2(5.6)
retired	0	1(4.0)	1(2.8)
not working (other)	2(18.2)	9(36.0)	11(30.6)
Parent has ACH			
n(%) yes	0	7(28.0)	7(19.4)

Percentages may not add to 100 due to rounding. ACH = achondroplasia; SD = standard deviation.

RESULTS

Demographic/Health Characteristics for Children of Parent Participants

Demographic/health characteristics for the children of parent participants are shown in Table 2.

- 30.6% of parent participants (n=11) had children aged 2 to <5 years with ACH, 36.1% of parents (n=13) had children aged 5 to <9 years, and 33.3% of parents (n=12) had children aged 9 to <12 years
- Nineteen children were female (52.8%) and 17 were male (47.2%)
- Twelve parents reported child's health status as "excellent" (33.3%), 14 reported as "very good"(38.9%), 7 parents (19.4%) reported as "good," and 3 parents (8.3%) reported as "fair"

Table 2. Demographic/health characteristics of children of parent participants

	Spain (n=11)	US (n=25)	Total (N=36)
Child age, n(%)			
2 to <5 years	5(45.5)	6(24.0)	11(30.6)
5 to <9 years	4(36.4)	9(36.0)	13(36.1)
9 to <12 years	2(18.2)	10(40.0)	12(33.3)
Child gender, n(%)			
female	7(63.6)	12(48.0)	19(52.8)
male	4(36.4)	13(52.0)	17(47.2)
Health status (parent-reported), n(%)			
excellent	3(27.3)	9(36.0)	12(33.3)
very good	3(27.3)	11(44.0)	14(38.9)
good	3(27.3)	4(16.0)	7(19.4)
fair	2(18.2)	1(4.0)	3(8.3)
Age/time diagnosed with ACH, n(%)			
in utero	9(81.8)	12(48.0)	21(58.3)
at birth	1(9.1)	4(16.0)	5(13.9)
<2 months of age	1(9.1)	2(8.0)	3(8.3)
2-6 months of age	0	5(20.0)	5(13.9)
unknown (adopted)	0	2(8.0)	2(5.6)

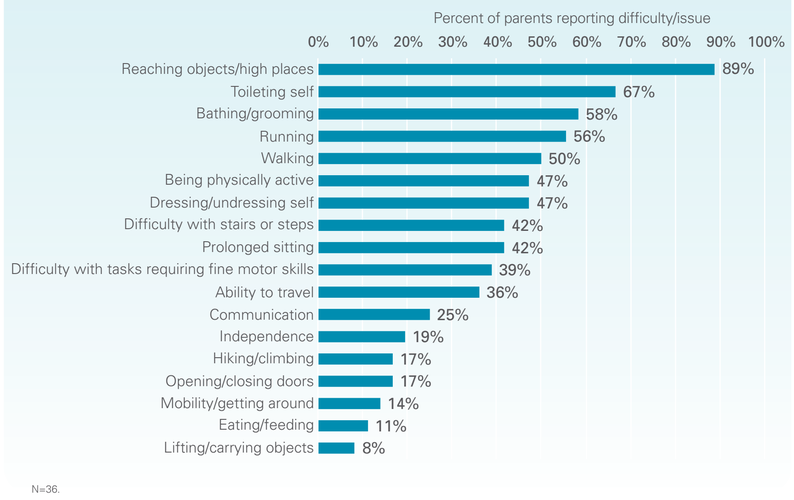
Percentages may not add to 100 due to rounding. ACH = achondroplasia; SD = standard deviation.

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 (56%, n=20)
- Walking (50%, n=18)
- Being physically active (47%, n=17)
- Dressing/undressing self (47%, n=17)

Figure 1. Impacts on children's functioning and daily life



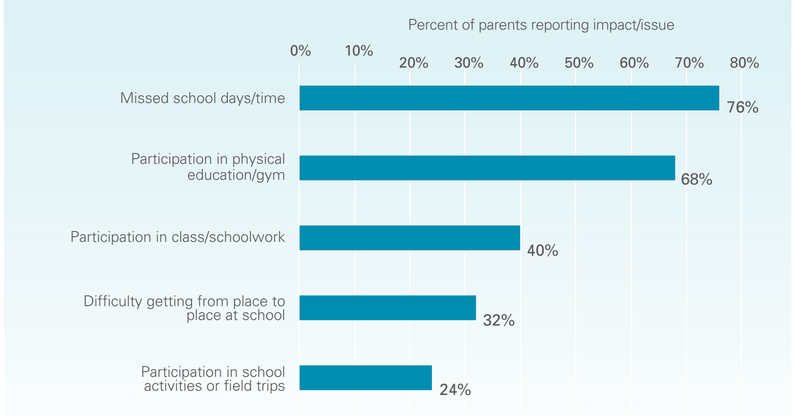
N=36.

Impacts on School Participation

Among school-aged children (aged 5 to <12 years, n=25), challenges to school participation included:

- Missed school time (76%, n=19)
- Limited/modified participation in physical education or "gym"class (68%, n=17)
- Difficulty participating in class/school work (40%, n=10)
- Difficulty getting from place to place at school (32%, n=8)

Figure 2. Impacts on children's school participation



Restricted to parents of children aged 5 to <12 years (n=25).

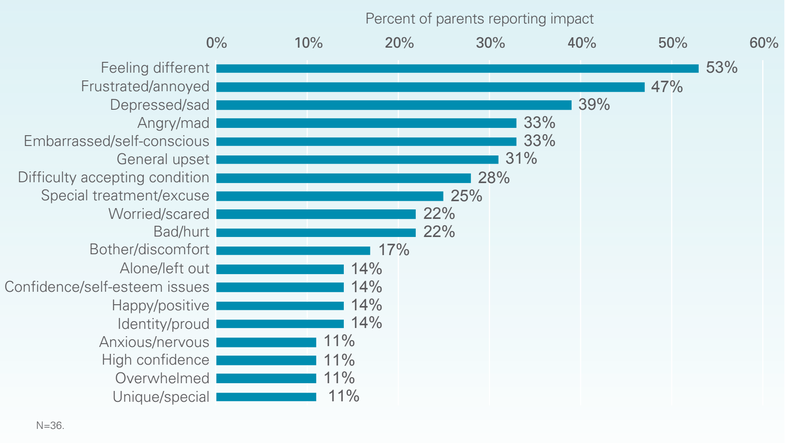
RESULTS

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/mad (33%, n=12)
- Embarrassed/self-conscious (33%, n=12)

Figure 3. Impacts on children's emotional well-being



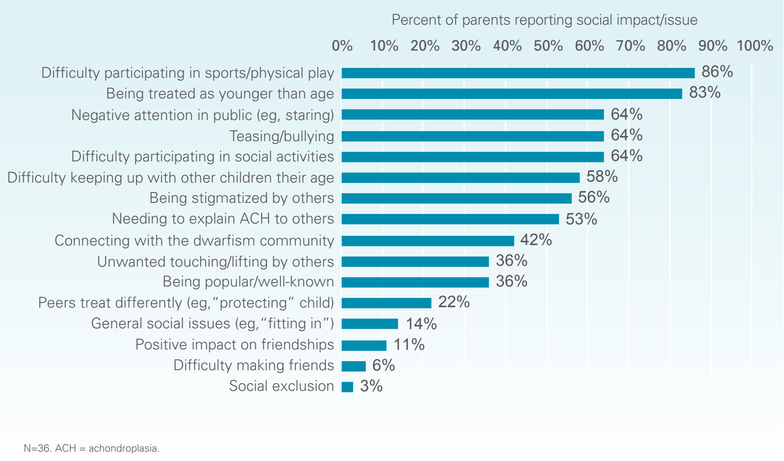
N=36.

Impacts on Social Well-being

The most frequently reported impacts on children's social well-being were:

- Difficulty participating in sports/physical play (86%, n=31)
- Being treated as younger than age (83%, n=30)
- Negative attention in public, such as staring/pointing (64%, n=23)
- Experience of teasing/bullying (64%, n=23)
- Difficulty participating in social activities, such as birthday parties/playdates (64%, n=23)

Figure 4. Impacts on children's social well-being



N=36. ACH = achondroplasia.

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

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