# **Development of a Parent Experience Measure for Parents of Children With Achondroplasia**

Kathryn M. Pfeiffer<sup>1</sup>, Meryl Brod<sup>1</sup>, Dorthe Viuff<sup>2</sup>, Sho Ota<sup>3</sup>, Jill Gianettoni<sup>3</sup>, Jonathan A. Leff<sup>3</sup> <sup>1</sup>The Brod Group, <sup>2</sup>Ascendis Pharma A/S, <sup>3</sup>Ascendis Pharma, Inc.

BACKGROUND
------------

- The clinical complications and medical impacts of achondroplasia (ACH) in children are well studied and frequently include recurrent ear infections, sleep apnea, hearing loss, teeth crowding, speech delay, and delayed developmental milestones, including gross motor and fine motor<sup>1–5</sup>
- Little is known about how having a child with ACH impacts parents' experiences and quality of life
- Research has shown that at the age of 7 years, many children with ACH continue to require minimal to moderate parent/ caregiver assistance with self-care, and some children still require supervision in social settings<sup>5</sup>
- Evidence also suggests that parents may experience emotional and other impacts at the time of their child's diagnosis<sup>6</sup>

OBJECTIVE

**Demographic/Health Characteristics for Children of Parent Participants** 

RESULTS

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

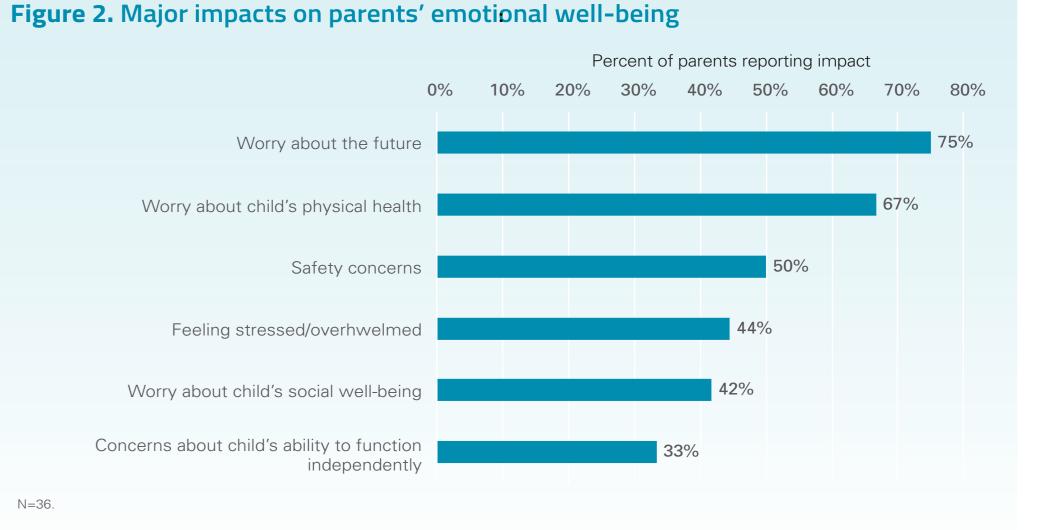
• Nineteen children (52.8%) were female and 17 (47.2%) were male

• Twelve parents (33.3%) reported child's health status as "excellent,"14 (38.9%) reported as "very good,"7 (19.4%) reported as "good," and 3 (8.3%) reported as "fair"

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



# RESULTS



The purpose of the study was to conduct concept elicitation interviews with parents of children with ACH to provide qualitative evidence to support the development of the Achondroplasia Parent Experience Measure (APEM), which assesses the impacts of having a child with ACH aged 2 to <12 years on parents' daily life and well-being.

# 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 (PROs).

Based on a literature review and clinical expert interviews, a semi-structured interview guide was developed to elicit parents' experiences related to having a child with ACH.

Concept elicitation sample inclusion criteria:

- an 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

Concept elicitation sample exclusion criteria:

• A cognitive impairment or other medical condition, including psychiatric conditions, that would affect a participant's ability to participate 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

	(n=11)	(n=25)	(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(%	6)		
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

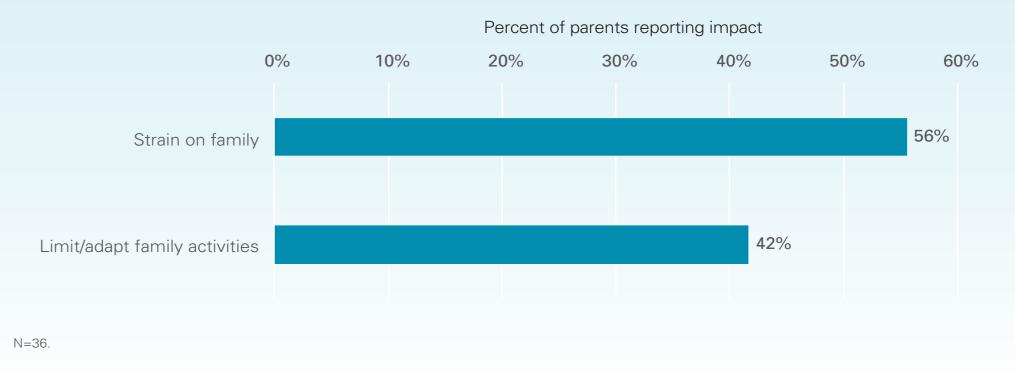
The qualitative analysis and the development of a preliminary theoretical model identified 4 conceptual domains for the impacts of having a child with ACH on parents, as well as the major impact(s) in each domain:

- Caretaking responsibilities:
- 6 major impacts (Figure 1)
- Emotional well-being:
- 6 major impacts (Figure 2)
- Family:
- 2 major impacts (Figure 3)
- Work:
- 1 major impact (Figure 4)

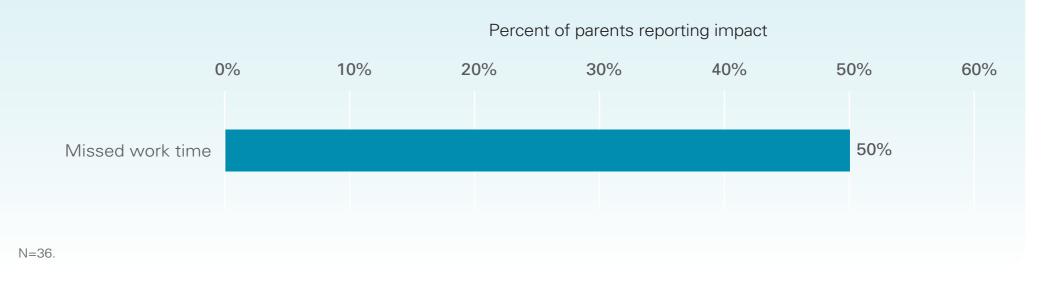
Preliminary theoretical model for Experience of Parents of children with achondroplasia (children ages 2 to <12 years)

	DOMAINS		Examples
	IMPACTS (PROXIMAL)		DISTAL IMPACTS
	Caretaking Responsibilities		Caretaking
MAJOR		MINOR	Responsibi
<ul> <li>Manage child's medial care</li> </ul>	•Observe child (eg,	<ul> <li>Obtain adaptations</li> </ul>	• Less overa
(eg, arrange/go to appts.)	to ensure child's	<ul> <li>Find childcare/school</li> </ul>	free-time
•Help child with self-care	safety, monitor child's symptoms like		
(eg, toileting, bathing,	sleep problems)		
dressing)	<ul> <li>Actively support child</li> </ul>	-	Emotional
Provide assistance to child	(eg, advocating for		Well-Being
(e.g. reaching objects, help getting from place	accommodations,		• Long-term
to place)	educating others)		worries
	•Provide support/guidance		
	to child about living with		Family
	and managing condition		Family
			• Family
	Emotional Wall being		stress/divo
	Emotional Well-being		• Impact on
MAJOR	MINOR	Proud	siblings
Concern about child's	<ul> <li>Worry about child's emotional well-being</li> </ul>	• Proud	
social relationships	-	• Feel normal	Work
• Worry about child's ability	<ul> <li>Acceptance of child with condition</li> </ul>	Feel protective of child	
to function independently	• Feel positive/focus on	Depressed/sad	• Take time
<ul> <li>Feel stressed (eg, overwhelmed)</li> </ul>	positive	• Feel hurt/bother	of workfor
• Concern about child's	Feel fortunate/lucky		
future	Increased knowledge		Social
•Worry about child's general	<ul> <li>Increased empathy/</li> </ul>		• Reduced/
physicalhealth	understanding		changed
• Worry about child's safety	• Overcoming challenges		social life
, , ,			
MAJOR	Family	MINOR	Physical
• Experience strain in family		<ul> <li>Increase family closeness</li> </ul>	• Decreased
•Need to limit/adapt family			overall ger
activities			health
	Work		
MAJOR		MINOR	Economic
• Missed work time (eg,		<ul> <li>Need to change work</li> </ul>	<ul> <li>Reduced</li> </ul>
arrive late, left		schedule	financial
early, or miss full day)		<ul> <li>Need to discontinue work</li> </ul>	status
			<ul> <li>Lost job</li> </ul>
	Social		opportunit
MAJOR		MINOR	
•None		<ul> <li>Limit social/other activities</li> </ul>	
		<ul> <li>Strained relationships</li> </ul>	
	Physical		
MAJOR	,	MINOR	
Nono		• Tired	
•None			





## Figure 4. Major impacts on parents' work



The newly developed APEM measure included 15 items in 4 conceptual domains.

- The APEM was designed as a patient-reported outcome (PRO) 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

## <12 years with ACH.

Interview and focus group transcripts were analyzed for content and coded by themes using a qualitative analysis software program.

The qualitative analysis report was used to develop a preliminary theoretical model of the impacts of having a child with ACH on parents and potential modifiers to inform the content and structure of the APEM measure.

Only impacts identified as major would be included in the measure.

## **Criteria for Identifying Major Impacts:**

- Endorsement of at least 30% of parent participants in at least 2 of the 3 child age groups analyzed; or an endorsement of 25% to 29% of parent participants in at least 2 of the 3 age groups if conceptually important
  - Endorsement percentages were considered across differing child age groups to ensure relevance to parents who have children of different ages
- Would be responsive to child's treatment
- Considered bothersome, limiting, or difficult
- Impacts must be proximal (rather than distal)

Once the APEM measure was developed, cognitive debriefing interviews were conducted with an additional 16 parents in the US to ensure that measure items were relevant and appropriate, and that instructions and items were easy to understand and complete.

# 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%)

# **POTENTIAL MODIFIERS** 1

<ul> <li>Child's age</li> <li>Parent/sibling achondroplasia status</li> <li>Socio-economic status</li> <li>Country</li> <li>Insurance coverage</li> <li>Coping strategies</li> <li>Healthcare system/ structure accessibility</li> </ul>	<ul> <li>HCP understanding/ knowledge about achondroplasia</li> <li>Severity of achondroplasia</li> <li>Treatment history</li> <li>Surgical interventions</li> <li>Number and/or severity of child's co-morbidities</li> <li>Child's siblings</li> <li>Degree of social support</li> </ul>	<ul> <li>Degree of family support</li> <li>Level of social acceptance</li> <li>Degree of accessibility in environment for people with achondroplasia</li> <li>School level of support/ accommodations provided</li> </ul>	<ul> <li>Parent's access to resources/ education about condition</li> <li>Parent type of employment</li> <li>Use of adaptive devices</li> </ul>
--	--	---	---

The conceptual framework for APEM is shown in Figure 5.

The APEM is a validation-ready PRO designed to assess the impacts of having a child with ACH aged 2 to <12 years on parents' general well-being, including caretaking responsibilities, emotional well-being, family, and work.

### Figure 5. Achondroplasia parent experience measure (APEM – Impact) conceptual framework

#### In the past 2 weeks, because your child has achondroplasia, how much time have you spent:

- Managing your child's medical care (such as arranging and going to appointments, making treatment decisions)
- Helping your child with self-care (such as toileting, bathing, dressing)
- Providing assistance to your child (such as reaching objects/high places, help getting from place to place)
- Observing your child (such as ensuring child's safety, monitoring child's symptoms such as sleep problems)
- Actively supporting your child (such as advocating for accommodations at school, educating others
- Providing support/guidance to your child about living with and managing their achondroplasia

#### In the past 2 weeks, because your child has achondroplasia, how often did **you** feel:

- Concerned about your child's social relationships
- Worried about your child's ability to function independently
- Stressed (such as feeling overwhelmed)
- Concerned about your child's future
- Worried about your child's general physical health
- Worried about your child's safety

#### In the past 2 weeks, because your child has achondroplasia, how often have you:

- Needed to adapt or limit family activities Experienced strain in the family
- In the past 2 weeks, because your child has achondroplasia, how often have you:
- Missed work time (arrived late, left early, or missed a full day)

👈 Family

# CONCLUSIONS

• The study provides evidence to support the content validity for the validation-ready APEM parent PRO measure to assess the impacts of having a child with ACH aged 2 to <12 years on



APEM – PRC

- Emotional Well-being

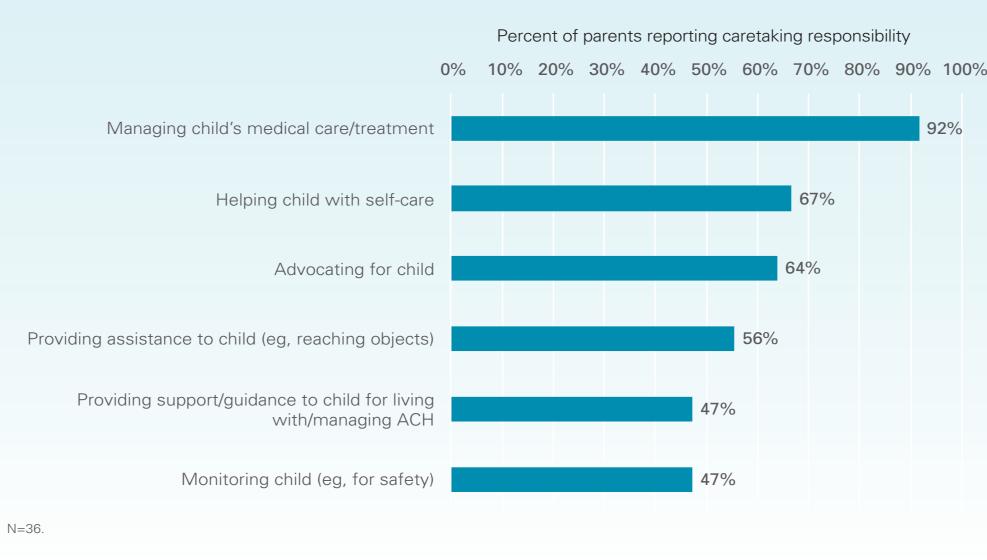
- Most participants were married (80.6%, n=29), 8.3% were partnered (n=3), 5.6% were divorced (n=2), and 5.6% were single (n=2)
- Seven parents (19.4%), all residing in the US, also had a diagnosis of ACH

## Table 1. Parent participant demographic characteristics

	<b>Spain</b> (n=11)	<b>US</b> (n=25)	<b>Total</b> (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

#### Figure 1. Major impacts on parents' caretaking responsibilities



parents' daily life and general well-being, including:

- Caretaking responsibilities (eg, managing child's medical care, helping child with self-care, assisting child, advocating for child, etc.);
- Emotional impacts (eg, worry about the future, worry about child's physical health, safety concerns, feeling stressed/ overwhelmed, worry about child's social well-being, etc.);
- Family strain (eg, having less time); and
- Missed work time to care for child
- A future psychometric validation study of the APEM is needed to further assess the measure's validity and reliability
- As new treatments for pediatric ACH are being developed, it is critical for clinicians to understand and assess the impacts of having a child with ACH on parents' lives, which may be lessened following children's treatment

1. Pauli RM. Achondroplasia: a comprehensive clinical review. Orphanet J Rare Dis. 2019;14(1):1

- 2. Hunter AG, Bankier A, Rogers JG, Sillence D, Scott CI, Jr. Medical complications of achondroplasia: a multicentre patient review. J Med Genet. 1998;35(9):705-712.
- 3. Wright MJ, Irving MD. Clinical management of achondroplasia. Arch Dis Child. 2012;97(2):129-134. 4. Ireland PJ, Donaghey S, McGill J, et al. Development in children with achondroplasia: a prospective clinical cohort
- study. Dev Med Child Neurol. 2012;54(6):532-537.
- 5. Ireland PJ, Johnson S, Donaghey S, et al. Developmental milestones in infants and young Australasian children with achondroplasia. J Dev Behav Pediatr. 2010;31(1):41-47.
- 6. Hill V, Sahhar M, Aitken M, Savarirayan R, Metcalfe S. Experiences at the time of diagnosis of parents who have a child with a bone dysplasia resulting in short stature. Am J Med Genet A. 2003;122A(2):100-107.





Growth and syndromes (to include Turner syndrome)

Poster presented at:

