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ORIGINAL RESEARCH

Treatment Goals for Achondroplasia: A Qualitative Study with Parents and Adults

Sarah A. McGraw · Jeff C. Henne · James Nutter · Adelpha Abrahamson Larkin · Er Chen

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ABSTRACT

Introduction: Achondroplasia is characterized by disproportionate short stature accompanied by other changes to the musculoskeletal system. Individuals with this condition typically experience a variety of medical complications. As pharmacologic treatments continue to be developed for the treatment of achondroplasia, it is important to understand treatment goals among those affected by achondroplasia and the factors that shape their goals.

Methods: This qualitative study is based on semi-structured interviews with 19 parents of children with achondroplasia and five adults with achondroplasia in the USA. We employed thematic analysis using an iterative process to identify themes across the interviews.

Results: Participants had two goals for pharmacologic treatment of achondroplasia: ameliorating complications associated with the condition and increasing stature to overcome functional limitations and psychosocial challenges. Complications of particular concern were chronic pain and surgeries to repair spinal, ear, nose, and throat (ENT) problems, and neurological sequelae. Increased height would enhance independence, help individuals to fit in socially, and avoid social stigma. Countervailing factors included the importance of stature to their identity and the concern that the condition would remain despite treatment.

Conclusions: This study offers evidence about how individuals affected by achondroplasia think about the pharmacologic treatment of this condition, including both the benefits of ameliorating complications and increasing height. The findings can offer practical insights for parents of children considering treatment, treating physicians, and decision-makers evaluating coverage decisions for treatment of achondroplasia.

Keywords: Achondroplasia; Treatment; Medical complications; Short stature; Social needs; Qualitative

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S. A. McGraw (✉) · J. Nutter
MNW Consulting Group, 4119 SE Bybee Boulevard, Portland, OR 97202, USA
e-mail: sarahmcgraw03@gmail.com

J. C. Henne
The Henne Group, 425 2nd Street, Suite 400, San Francisco, CA 94107, USA

A. A. Larkin · E. Chen
BioMarin Pharmaceutical, Inc., 770 Lindaro Street, San Rafael, CA 94901, USA

Key Summary Points

Why carry out this study?

Achondroplasia, a genetic disorder that inhibits endochondral bone growth, is characterized by disproportionate short stature, other changes in the musculoskeletal system, and significant medical complications.

Until recently, surgery was the only treatment commonly available to address short stature and shortened limbs but new pharmacologic therapies are emerging, and little is known about how patients and their families view the use of these therapies.

This exploratory study sought to understand the treatment goals of caregivers of children with achondroplasia and adults with the condition.

What was learned from the study?

An increase in stature to allow independence, minimize stigma, improve social life, and preserve their quality of life into adulthood is important to many participants, while prevention of disease-related complications is also a major driver for treatment.

Treatment choices require weighing the benefit of achieving desired outcomes and, for some individuals, the potential for losing their identity as a little person.

This new information can help to guide physicians caring for this population as well as policies regarding coverage of treatment options.

INTRODUCTION

Achondroplasia, a skeletal dysplasia, is the most common form of disproportionate short stature [1, 2]. It is an autosomal dominant condition caused by a pathogenic gain of function mutation in the fibroblast growth factor receptor 3 (*FGFR3*) gene, leading to an inhibition of endochondral bone growth [1–4]. Achondroplasia is a rare condition, occurring in approximately 1 in 25,000 births [3, 5]. An estimated 80% of cases are the result of a spontaneous pathogenic variant and most individuals are born to parents of average stature [3]. Occurrence increases with paternal age [3]. Mortality rates are higher in this population across all ages, and average life spans are shorter [6].

The physical features of achondroplasia are characterized by changes in the musculoskeletal system that include short stature, shortened proximal section of limbs (rhizomelic shortening), macrocephaly, frontal bossing with midface hypoplasia, and atypical development of the mandible [7, 8]. In addition, individuals with this condition may have muscular hypotonia and kyphosis of the spine in infancy, and then lordosis when they begin walking. They may also develop a smaller than normal chest size; shortened fingers with a trident hand-shape; hypermobility of hips; and leg bowing [2].

Secondary medical complications, some of which can be life-threatening or have serious health consequences, are associated with achondroplasia. They emerge in infancy, and many continue into adulthood [9, 10]. Unger et al. [11] grouped complications into four areas: (1) ENT problems including hearing loss; (2) sleep apnea and other pulmonary-related concerns; (3) neurologic problems; and (4) orthopedic problems [1, 12]. Problems tend to occur in predictable age groups and age-specific surveillance is recommended [1, 2, 11, 12]. Complications associated with achondroplasia can require multiple surgeries over the lifetime [13]. Chronic pain is common and increases with age [14, 15].

Functional limitations can result from the various physical features and complications associated with achondroplasia. Delays in motor development are associated with anatomic differences such as disproportionately short limbs, stiff elbow, hypermobility of other

joints, and macrocephaly [2]. Children need assistance with self-care including perineal hygiene, grooming, bathing, and dressing [11]. Other functional limitations include difficulties with locomotion such as climbing stairs, transferring on and off chairs, walking or running, and sitting in chairs without leg support [16, 17]. These challenges lead children with this condition to achieve independence, on average, later than non-affected children [18]. Difficulties around social engagement, emotional well-being, and school participation can arise [14, 16]. Pfeiffer [17] notes the importance of finding adaptations or assistance in the home and at school. In adulthood, people with achondroplasia may face limited educational and career choices, despite normal intellectual capacities [19].

Children and adults with achondroplasia can experience a lower quality of life (QOL) than their counterparts without this condition [17, 19–23]. QOL is influenced by several factors including disease severity, height, pain, age, self-esteem, social support, and perceived stigmatization [14, 19, 20, 24]. Several studies, however, did find that while QOL scores related to the physical function domain were lower than in reference populations, mental or emotional components scores were not [22, 23].

Concerns about medical complications, functional limitations, and overall QOL have led to an interest in the treatment of this condition. Procedures to manage secondary complications (e.g., suboccipital decompression for craniocervical compression; positive airway pressure for sleep apnea; placement of ear tubes for middle ear dysfunction) have been in use [2]. To date, the most commonly used approach to alter to short stature or disproportionate limbs has been limb lengthening which involves multiple surgeries and prolonged recovery [12, 25]. As an understanding of the pathogenesis of achondroplasia improves, attention is turning to pharmacologic treatments [7, 26, 27]. While the potential benefit of pharmacologic treatment is documented in the clinical literature, little is understood about how patients and families view the use of medications to improve endochondral bone growth to increase their stature and, potentially, ameliorate or prevent complications.

The purpose of this exploratory study is to begin to address this knowledge gap. We seek to understand the treatment goals of parents of children with achondroplasia and adults with the condition and the factors that shape their goals.

METHODS

Study Design

For this qualitative study, we report on semi-structured interviews with 19 parents of children with achondroplasia and five adults living with the condition. We used thematic analysis to examine the interview transcripts [28, 29]. All participants signed an informed consent document before the interview with a description of the study aims and interview topics. The conduct of this study followed the COREQ criteria [30]. Ethics approval for this study was obtained from the Ethical & Independent Review Services (IRB00007807). Informed consent was obtained from all study participants and all participants gave written consent to use study data in publications, including use of unidentified excerpts from their interviews. Study procedures were conducted in accordance with the Helsinki Declaration of 1964 and its later amendments.

Recruitment

Since the goal of this qualitative study was to explore the views and experiences of people living with this rare condition, we used purposive sampling to recruit parents and adults from a range of age groups [31]. As a result of the rare nature of the condition, we aimed to recruit enough participants in both groups to allow us to reach content saturation.

Parents and adults with achondroplasia were recruited through patient advocacy groups such as Little People of America and a list of individuals from a website (achondroplasia.com) who agreed to be contacted for research activities. Recruitment materials described the

Table 1 Number of individuals: recruitment, eligibility, and interview completion

	Number
Sent study materials	161
Completed screener online	48
Ineligible	
Living outside of USA	6
Enrolled in other studies	6
Declined invitation	4
Eligibility not verified	1
Completed screener after quotas filled	5
Total Ineligible	22
Invited to participate	26
No response to invitation	2
Completed interview	24

research firm conducting the study and named the pharmaceutical firm which sponsored the study. We emailed these materials to potential participants, inviting them to complete a study screener to determine eligibility. Parents were eligible to participate in the study if they had a living child with achondroplasia who was under the age of 18. Adults aged 18 years or older and diagnosed with achondroplasia were also eligible. The exclusion criterion was participation in an achondroplasia clinical trial in the preceding 12 months. Table 1 shows the number of individuals at each phase of recruitment and enrollment. A total of 24 parents and adults completed interviews.

Data Collection

Two interviewers (McGraw and Henne), both lead researchers at their respective research firms, completed the semi-structured interviews. Dr. McGraw (female) holds a Ph.D. in medical anthropology and Mr. Henne (male) an M.A. and each has over 25 years in qualitative research. Neither interviewer had contact with the study participants before the interviews. At the outset of the interview, they explained that they were conducting the study as professional interviewers and did not have a medical background or in-depth knowledge about achondroplasia.

The interview questions for parents and adults covered their background and family life; how achondroplasia affected their lives including the social and functional effects of the condition; the complications they experienced, primary concerns for themselves or their child; their goals for treatment; and the perceived benefits of treatment (see the electronic supplementary material for the interview guides). Interviews lasted 60 min. The interviews were conducted and recorded through an online meeting space. In addition to the interviewer and the participant, a member of the study staff was in the background to manage the meeting space and recording. Identifying information was erased from the recording before verbatim transcription. Upon completing an interview, participants received US $75.

Analysis

Thematic analysis followed an iterative and a consensual process [28, 29, 32, 33]. Deductive or a priori codes were informed by an earlier internal study (unpublished, 2021). Inductive or open codes were identified through an initial reading of the transcripts. To begin coding, a lead coder (SM) read and identified salient concepts for each transcript. Second, two members of the coding team (SM and JN) independently read the same set of ten transcripts. A third coder served as an auditor, reviewing a subset of transcripts. After each set of transcripts was coded, the team met to reconcile differences, revise the code list, and recode (see Table S1 in the electronic supplementary material for the list). Joint coding and reconciliation broadened the capture of salient constructs while ensuring the codes were consistently applied. Following an iterative process, team members reviewed and synthesized the text for each code to identify patterns and summarize underlying concepts [29]. They

grouped the concepts into higher-order categories, identifying cross-cutting themes [31]. NVivo was used to facilitate coding and analysis [34]. Themes were tracked by individual participants throughout the study to monitor thematic saturation [35].

To enhance the readability of verbatim quotes, we removed verbal hesitations or repetitions. Quotations in the text are identified with a number to indicate the speaker's participant group: C for parent and I for adult.

RESULTS

Study Population

The study population included 19 parents who had a child with achondroplasia and five adults with this condition. The parents were predominantly female; white; had college or postgraduate education, and worked full-time (Table 1). None of the parents had this condition themselves. All respondents in the parent cohort had only one child with achondroplasia. Their children ranged in age from 1 to 13 years; 10 were 5–12 years old. Ten of their children were male.

The five adults with achondroplasia ranged in age from 27 to 67 years; three were female; and all were white. Four had a college education or more, and three were employed (Table 2). Three of these adult participants had a child (or children) and/or a spouse with achondroplasia. Four adults mentioned that their parents were of average stature.

Table 2 Characteristics of parents of children and adults with achondroplasia

	Parents (*N* = 19)	**Adults (*N* = 5)**
Gender		
Female	16 (84)	3 (60)
Male	3 (16)	2 (40)
Ethnicity		
White	16 (84)	5 (100)
Black	1 (5)	0
Latinx	2 (11)	0
API	0	0
Education		
Some high school or GED	3 (16)	0
Some college/technical	2 (11)	1 (20)
Four-year degree	6 (32)	3 (60)
Postgraduate	8 (42)	1 (20)
Employment status		
Full-time	11 (58)	3 (60)
Part-time	5 (27)	0
Homemaker	1 (5)	0
Unemployed	2 (11)	1 (20)
Retired	0	1 (20)
Others in family with achondroplasia[a]		
Partner	0	3
Sibling	0	1
Children	0	3

Data are presented as *n* (%)

[a]3 of the 5 adults with achondroplasia had others in their family with the condition

Themes About Treatment Goals

We organized the participant's description of their experiences with the condition and their treatment goals into three thematic areas: (1) pharmacologic treatment can ease their lives through increased stature; (2) treatment to ameliorate or prevent complications was important; and (3) the presence of countervailing views about the benefits of treatment.

Benefits of Increasing Height

Increasing stature was not the primary outcome for the study participants; rather, increasing

stature could help in three ways: (1) creating an independent life in an average height world by removing functional limitations; (2) fitting in socially; and (3) minimizing discrimination and stigma. Representative quotations about the benefits of pharmacologic treatment for each area are shown in Table 3.

Improving Function and Independence in an Average Height World

Functional limitations related to short stature had a broad effect on the lives of people with achondroplasia both inside and outside the home. Reaching countertops or shelves in stores, handles of shopping carts, pushing elevator buttons, or using ATMs could be difficult. Using a toilet, sink, or shower faucet required a stool or modification of bathroom fixtures. Stair risers could be too high to climb or doors hard to push open and walking long distances tiring. Finally, carrying large or heavy items such as a backpack or even carrying a child presented challenges. These activities were difficult because, as a parent said, "everything in this world is designed for that certain height" (C01). Increased stature would ease the performance of these daily tasks, impacting "every moment of the day" (C20) and supporting greater independence.

It is important to note that while participants described the challenges of short stature and the benefits of increased stature, all of them found ways to handle their functional limitations through adaptive measures. As one adult said: "You learn to especially when it comes to things with school or even in the real-world setting, like how do I brush my teeth or things like that, you find solutions" (I06). Another adult commented that "we can do absolutely anything anyone else can do. It just might be harder" (I10).

Fitting in Socially

Differing in appearance from people of average stature created challenges in forming friendships and finding life partners. A mother observed: "So it's difficult for the people who have physical differences to make friendships or get in love or something like that" (C17). Another believed that it was difficult for her child to "be fully embraced and… accepted" (C18). Short stature presented challenges to everyday social interactions. For example, height differences can make it difficult to have face-to-face interactions with people of average stature.

As suggested by the quotations in Table 3, increased stature could help by making the appearance of people with achondroplasia more like that of people of average height. Increased stature could limit potentially uncomfortable social interactions and help in forming relationships. By addressing concerns about fitting in socially, increased height might also bring psychological benefits.

Avoiding Stigma and Discrimination

Participants explained how physical differences and the necessary use of aides to function triggered experiences of stigma and even discrimination. They described situations in which they, or their child, were exposed to inappropriate attention such as staring, pointing, being photographed without permission, or name-calling. Standing out or being different in school was difficult for children with achondroplasia. A mother described her daughter's embarrassment about needing adaptive devices to use the toilet in school because it drew unwanted attention and made her feel stigmatized. She reported that her daughter said:

> "I don't like having to have a stepping stool every time I go to use the restroom." Or, "I don't like having to have to carry these wipes with me when I use the bathroom because people ask questions." [She says], "Sometimes I just hate it when people ask or point at me… What are you pointing at?" (C33).

Finally, participants described instances when they believed that they were denied jobs or rejected from training programs on the basis of their short stature. Parents of children with achondroplasia and adults believed that with increased stature they might avoid these problems.

Table 3 Illustrative quotes related to the benefits of increased stature

Theme	Representative quotations
Function	
Toileting and personal hygiene	She's going to be a little person who maybe doesn't need any accommodations in a public restroom. She's not going to have to lift herself up onto a toilet, or she's going to be able to reach a sink (C20)
	[Ease] feminine hygiene (C16)
Dressing	It would be about height, the fact that she could do her own hair without having to fight with me about the way I styled her hair, about being able to take a shower without having to say, 'Mom, can you help me here?' (C33)
	I would love to see her be five feet tall with average arm span as well with that height. Because I think that would… give her some space to maybe be able to find some clothes, maybe (C14)
Reaching	I want her to have an independent, high-quality life. And if that means being able to reach something on the shelves when she goes to the grocery store without asking somebody because it's on the third shelf and she can't reach it, great. I don't think getting height is a bad thing (C32)
	Being able to reach the door handle, being able to turn on the light (C14)
	Access an ATM or reach a counter or be able to, then I don't see how [increased stature] could be a negative (C22)
Walking longer distances	'I just want to be able to walk across campus to my own classes.' That was her reason for doing [limb lengthening]. She went to this state college that has a beautiful campus, that like part of the joy of the school is walking across this beautiful campus to go to your classes, and that's what she wanted (C20)
Driving	If you gain a few inches, maybe you can drive a car without having all the equipment. Reach out to a door opening and opening a window (C12)
Athletics and play	[She] 'could have ridden [a bike] when she was little with all the other kids or gotten to do the bounce houses' (C31)
Safety	Be visible by vehicles when they're driving by (C22)
Access	They're never going to lose the things that make them unique, but it is going to give them an ability to be accessible to the world (C22)
Fitting in	
Looking like others	But I think for some parents, especially when their children are very young, there's a lot of anxiety about how their child's going to be different, how can they make them so they're not different. And so, when we talk about treatments for stature and that sort of thing, they're often the ones that are pushing that approach (P23)
Removing social discomfort	One of the things she said was when she would speak to someone… she was eye level to their crotch. I'm not a little person, so I wouldn't even think of that. And I'm like, 'Oh my gosh.' Like she would always have her head down. She would never look up (C26)

Table 3 continued

Theme	Representative quotations
Forming relationships	Maybe it would have been easier socially to make friends. And if I wanted to marry an average sized person, it probably would have made that easier. So, I can definitely see how it could have made life easier (I10)
General psychological benefits	The social, emotional, mental health benefits to her would be big… [because she would] stand out less from her peers (C16)
	If [increased stature] would take away… social issues, psychological, emotional issues, then I'd say that it's probably a positive. I think everyone probably felt that way about one drug that came out over another and how it's going to change your quality of life (C22)
Stigma and discrimination	
Job discrimination	Or she can walk in for a job interview [where they ask] 'Oh, will she be able to do the job?' I think it will be able to give her more of an identity of her as a person and not achondroplasia (C31)
	She would be less likely to face potential discrimination due to her size if she was able to have more growth (C16)
Bullying	Closer to at least the bottom range of the average height people so that he doesn't stand out. People don't always point at him and… bully him (C21)

Ameliorating or Preventing Complications

Preventing or ameliorating complications was important to all participants and they hoped that pharmacologic treatment would offer this benefit. The four areas of potential benefit were prominent: spinal problems, pain, surgery, and problems with aging. See Table 4 for representative quotations.

All but one of the participants described repetitive ENT problems leading to the need for tube placement as well as potential hearing impairment. More than half of the participants hoped that treatment would lessen foramen magnum stenosis, the most life-threatening complication. Chronic pain affected daily lives and limited physical activities or social participation. A mother said that her son "was going to the nurse to get [painkiller] during the day" (C25), and she assumed that these visits limited his classroom time. They hoped that pharmacologic treatment would keep their children from experiencing pain, including joint pain, and pain from surgeries. Both parents and affected adults mentioned one or more surgeries for a variety of problems including ear tube placements, tonsillectomies, spinal decompression, orthopedic surgery for leg bowing, or surgery for spinal stenosis. In many instances, these surgeries were required at a very young age. In commenting on the value of treatment, a mother exclaimed that if a treatment had been available that could prevent many of the complications her child experienced, she would have started treatment as soon as her daughter was born.

> "She had decompression surgery. She's had four sets of tubes. She's had her adenoids out twice. If that all could be avoided, yep. If somebody had said to me at 33 weeks pregnant, 'the day your baby is born, we can give them medicine that could help with avoiding all of that', I would have started day one. I would not have hesitated" (C31).

Table 4 Illustrative quotes related to ameliorating or preventing complications

Theme	Representative quotations
ENT problems	So, they have a lot of airway issues. I feel like that wouldn't be as prevalent. The sleep apnea wouldn't be as prevalent (C22)
	Maybe if they get the treatment, they wouldn't need ear tubes. Yeah, I definitely think there's some benefits (I01)
Spinal problems	… if they had come to me and said, 'Oh, this medication is going to widen her foramen magnum and… she's not going to need a neck surgery or shunt surgery', of course, that would be something that my husband and I would have totally talked about and considered (C9)
	'Anything that can prevent issues with his spine, those are very strong selling points for me' (C27)
Surgery	I think that his life would be tremendously better. He would never have to worry about getting a hip surgery or leg straightening or anything like that. To know that you don't have the major surgery on the horizon I think is a pretty good thing (C22)
	But if there was something that would make it so my child didn't have to have surgery on her legs someday or spinal cord decompression surgery and it's as simple as like a little prick with a needle, yes, I'd be open to it and learning more (C11)
Pain	That there wouldn't be the back pain, the leg pain, the knee pain, the neck pain, the surgeries… that would be the most important (C14)
	I think the large majority of achondroplasia individuals are going to be motivated by reducing more of the physical pain and hospitalization surgeries (I06)
Problems in adulthood	I want to decrease the chances that my kid's going to be using a walker in her late fifties or have surgeries or pain or anything (C20)

Countervailing or Moderating Views

Participants expressed countervailing views or questions about the benefits that pharmacological treatment could offer patients. They had three concerns: (1) despite treatment, a patient would still have achondroplasia; (2) people with achondroplasia valued their identity as little people and question the importance of increasing stature; and (3) how to weigh the value of increasing height compared to preventing complications. Quotations illustrating each concern are shown in Table 5.

Achondroplasia Remains After Treatment to Increase Height

While recognizing that treatment could increase stature, they anticipated that it would not change the fact that their child has achondroplasia, or as one parent said they will "still have the gene" (C09). Parents also suggested that treatment may not address other phenotypic features of the condition: "Sure, you're getting some height, but you're not really addressing a lot of the other phenotype characteristics. Like, the trident hand, your feet, the larger head, and frontal embossing" (I06).

Height is Part of Our Identity and There is Nothing Wrong with Being Little

Being a little person was an important part of identity for many and they liked who they were as little people. One parent explained simply that their child, "likes being small" (C12). An adult described how his experiences living with achondroplasia helped him to see the world differently, and be more creative. He said: "I

Table 5 Illustrative quotes related to countervailing views

Theme	Representative quotations
Achondroplasia remains after treatment	I'm just trying to be a realist. I don't think it really will change much how he looks because achondroplasia is a condition that is the only dwarfism that is disproportional. Like your head, a regular-sized trunk, but shorter limbs. They look different, and I don't know how much really will change that. And also, adding the fact that he has hydrocephalus. His head is even larger (C27)
	Increasing height can be a little nicer just because your capabilities might improve slightly. But without improving the everything, like hands, leg—or your hands, feet, and the other characteristics, then you still aren't—you're still living with the condition, the appearance, and that type of thing (I06)
Short stature is part of our identity	I think people just need to realize that just because someone is short in stature, that's it… There's no cognitive delays or things like that. So, I think it's just a matter of people becoming familiar with achondroplasia and with dwarfism in general and just treating people like people and not be so affected by the height difference (C14)
Weighing benefits of increasing stature to preventing complications	I think that would be easier… I lump the medical complications with having to have corrective long bone surgery to correct bowed legs, or spinal stenosis, or brain and spinal surgeries… That's a different worry or concern than he can't reach the gas pump, or he can't reach door handles. Like, we can figure that out, we can help him with that, and that is not such a big deal to us anymore. And to him (C28)
	I think the reduced risk of complications, of the back surgeries and such. Just reduce the risks of all the infections and everything. Height is only what you make it. Beauty is only skin deep. It's how we see it (I15)

think a lot of that has actually led to my creativity, which has helped me from a professional standpoint of always looking at problems from a different lens" (I06).

The suggestion that they would want to increase their height was seen by some as being offensive and dismissive. In their view, the world should learn to be more accepting of them. One parent said: "[Little people] kind of want society to be better instead of—they don't think they should try and change for society, and that's kind of how we feel" (C09).

Relative Benefits of Increasing Stature and Addressing Complications

Some participants acknowledged that while increasing stature might be helpful, they cared more about addressing complications. They would be less interested in the treatment if it only increased stature because many of the functional limitations or social concerns can be addressed using adaptive devices or learning how to cope with social situations. In the view of these participants, the medical complications should be the primary focus of treatment. As one mother commented:

> "Not just the height. That is not something that I really care about. I care more about [whether] it will improve the size of the thoracic cavity and the shape of the facial structure and the foramen magnum, because there are some kids that have a recurring narrowing of the foramen

magnum even though they had already had the surgery done. So, if those are positive things that that drug can solve, I will consider it" (C27).

DISCUSSION

Our study showed that treatment goals with potential pharmacologic agents centered around two features of life with achondroplasia: (1) medical complications and (2) limitations to physical function and social adaptation due to short stature. While the benefits of addressing both features were apparent to participants, doubts and some negative views also were noted, particularly the importance of identity as a little person.

The challenge of functioning in a world built for people of average stature, experiences with stigma, and the effects on their QOL have been described in other literature, but this is the first study, to our knowledge, to explore how their experiences can inform treatment goals [14, 17, 19, 20, 22, 36]. They saw benefits to increased height related to increasing one's stature in terms of increased accessibility, independence, and acceptance but they wanted to value and honor their identity as little people.

Addressing medical complications was the most important goal for some. While participants hope the pharmacologic treatment will offset complications and hope treatment may also improve some phenotypical features such as frontal embossing and larger heads, there is currently no clinical evidence in the scientific literature about these outcomes; however, such studies are underway [37, 38].

Our study did not set out to assess QOL, rather we focused on understanding how people affected by achondroplasia think about their experiences and how they perceive whether they may or may not benefit from treatment. Our findings do point to the importance of going beyond a single QOL measure to understand the range of experiences and views of persons affected by achondroplasia. Stature and complications are two separable domains of life experience and should be treated as such in measures to assess the effect of treatment on measures of QOL. This finding is consistent with the results of the concept elicitation reported by Pfeiffer et al. [17].

Some QOL studies indicate that not all individuals affected by achondroplasia have had negative life experiences [21–23]. Our findings support this. While our participants did face medical complications and physical limitations, positive views about identity as a person with achondroplasia were evident. Being of shorter stature is part of their identity; and for the most part, they are happy with that, and want to be accepted and valued as little people. In general, children are encouraged and supported by their parents to learn to accept themselves and their challenges. This finding might help to explain why Witt et al. [22] found that children with achondroplasia did not have lower scores on the emotional domain of QOL compared to their counterparts. It is possible that the children in this study arrived at self-acceptance and were happy with themselves.

Consistent with the literature on QOL, we found that having achondroplasia does affect their function and social experiences [17]. Social challenges such as stigma and discrimination were a concern for study participants. Such experiences can affect a sense of well-being and are consistent with the items suggested by Pfeiffer [17] for the Achondroplasia Child Experience Measure (ACEM). While many individuals found ways to adapt to an average height world, some continued to experience difficulties with functional limitations. Increased height might help to address some of these challenges.

Future studies might examine the degree to which the relative values placed on increased stature and preventing medical complications are mediated by identity as a little person and connection with the broader achondroplasia community. Another topic for future study is to assess the relationship between the severity of complications, degree of functional limitations, QOL, and treatment preferences. Both studies would benefit from a quantitative design with a larger sample.

Limitations and Strengths

Like many qualitative studies, the sample was small and not designed to represent the full population. The study participants were largely white, employed, and highly educated. As such, they may have greater resources to adapt to functional limitations. This study only draws on participants in the USA who were affiliated with patient groups or actively seeking information about achondroplasia. It is possible that participants who were aware that a pharmaceutical firm funded the study were more likely to express positive beliefs about pharmaceutical treatment. On the other hand, strongly held views against pharmaceutical intervention are held by some members of the achondroplasia community, and awareness of these attitudes might have counteracted any potential for a positive bias in response to the source of study funding. In addition, the interview guide was constructed to avoid leading questions about treatment preferences to minimize response bias; either negative or positive. One strength of the study sample was the inclusion of views from two sets of stakeholders in the achondroplasia community—parents and adults; however, despite our best attempt, we were not successful in recruiting adolescents whose views may differ from those of parents. While some of the views expressed by the participants in this study may not be broadly representative of the general population of people living with achondroplasia, they do offer insights into how parents and adults think about treatment and the factors that help to shape their views. The results suggest hypotheses that can be explored in future quantitative studies.

CONCLUSIONS

The study findings describe the spectrum of views about the benefit of pharmacologic treatment of achondroplasia. The results offer practical insights for parents of children with achondroplasia, physicians treating these patients as well as decision-makers evaluating coverage decisions for achondroplasia treatment. Parents may be more receptive to the benefits of treatment early in a child's life when they have the highest level of concern about complications and what the future will bring for themselves and their children.

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from all study participants. All participants gave written consent to use study data in publications, including use of unidentified excerpts from their interviews. Study procedures were conducted in accordance with the Helsinki Declaration of 1964 and its later amendments.

Data Availability. Data supporting the findings in the manuscript are included within the manuscript. Further data is available from the corresponding author.

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