RESEARCH PRIORITY REPORT

Convening to Identify Patient-Centered CER (Comparative Clinical Effectiveness Research) Priorities of Individuals with Achondroplasia and Their Families

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Introduction

Achondroplasia is the most common skeletal dysplasia resulting in short stature (dwarfism), affecting approximately 1 in 25,000 individuals.[1] While achondroplasia is genetic, caused by a change in the fibroblast growth factor receptor 3 (*FGFR3*) gene, approximately 80% of individuals with achondroplasia are born to average-stature parents with no personal or family history of the condition.[1]

Adults with achondroplasia reach a height of approximately 4 feet and typically use physical accommodations to navigate height-related differences.[2] Individuals with achondroplasia have an increased risk of developing health complications such as spinal stenosis, obstructive sleep apnea, lower extremity bowing, and recurrent ear infections, which may require medical or surgical interventions.[1,3] Until recently, individuals with achondroplasia who wished to pursue interventions to increase linear growth underwent extended limb lengthening (ELL), a procedure originally developed to correct limb length discrepancies.[2] In the United States, ELL is not considered standard of care for individuals with achondroplasia and is associated with significant complications, including fractures.[2,4]

In 2021, the FDA approved vosoritide (brand name VOXZOGO), a once-daily subcutaneous injection, to increase linear growth in children with achondroplasia.[5] Vosoritide is designed to counteract the altered FGFR3 signaling in children with achondroplasia who have open growth plates. Clinical trials have demonstrated an average increase in height of 1.57 cm/year (0.62 inches/year) sustained over 4 years; however, it remains uncertain whether this increased growth will result in greater final adult height or simply accelerate the rate at which typical adult height is achieved.[6,7] Some researchers hypothesize that vosoritide may also reduce the incidence or severity of certain health complications commonly associated with achondroplasia.[3] These potential benefits are an important area of interest for many families considering

pharmaceutical treatment options. However, there is currently a lack of robust clinical data to support that vosoritide use may lead to long-term medical benefits.[8] Moreover, publications hypothesizing potential benefits are often supported by pharmaceutical industry funding or collaboration, which may introduce conflicts of interest that influence how these findings are interpreted or presented.[9,10] Since the initiation of this PCORI-funded project, a second medication, navepegritide (brand name YUVIWEL), a once-weekly subcutaneous injection, has also been approved by the FDA to increase linear growth in children age 2 years and older with achondroplasia.[11]

Perspectives within the achondroplasia community regarding pharmaceutical therapies intended to increase height are diverse and nuanced. Some families pursue these treatments in hopes of increasing height and/or reducing medical complications. Others emphasize that short stature is not inherently a medical problem, but rather an essential component of individual and community identity.

Little People of America (LPA), the largest advocacy organization for individuals with dwarfism in the United States, has expressed apprehension about the emphasis of height as a primary treatment endpoint. In its official position statement, LPA described vosoritide as "a pharmaceutical solution for a societal problem," highlighting concerns about framing short stature primarily as a medical issue in need of correction.[11] This perspective aligns with the social model of disability, which emphasizes that it is not variation in individual bodies, but societal barriers, such as inaccessibility, stigma, and discrimination, that limit full participation and inclusion of disabled individuals, including those with achondroplasia.[12]

Community perspectives are often absent or underrepresented in medical literature about achondroplasia, including the recent *International Consensus Guidelines on the Implementation and Monitoring of Vosoritide Therapy in Individuals with Achondroplasia*.[10] The guideline development group did not include representatives from the broader achondroplasia community who chose not to pursue height-altering interventions, and concerns were raised regarding financial relationships with the drug manufacturer. Further, the guidelines did not address the range of lived experiences and quality of life considerations such as identity formation, emotional and psychological well-being, and community belonging. These considerations have been identified by advocates as vital for parents weighing treatment options for their children with achondroplasia.[13] Greater inclusion of community perspectives in research and guideline development is essential for families to access balanced, comprehensive information and make truly informed decisions.

Project Goal

The primary goal of this project is to identify, document, and disseminate the top patient-centered comparative clinical effectiveness research (CER) topics and research priorities of the achondroplasia community, while strengthening meaningful community engagement in research. It brings together adults with achondroplasia, parents of children with achondroplasia (including parents with and without achondroplasia), clinicians, and researchers. This approach is designed to ensure diverse representation of a broad range of community and professional priorities through facilitated conversations.

This project is crucial because perceptions about quality of life and care priorities of individuals with achondroplasia are often informed by research involving a relatively small subset of participants enrolled in pharmaceutical trials in which the primary endpoint is increased height. Quality of life research is typically conducted with and funded by the pharmaceutical industry, which may bias the research and distort broader community experiences and perspectives.[14,15] This project intentionally includes individuals with varied experiences, such as those who have pursued pharmaceutical interventions as well as those who have pursued medical, social, and community-based supports and services. In doing so, the project seeks to support a more comprehensive, balanced, and inclusive research agenda.

Methods

Aim 1: Identify three community-driven research priorities of primary importance by hosting an online conversation with individuals with achondroplasia and family members.

The purpose of this aim was to ensure that the voices, values, and priorities of people with achondroplasia and their families are documented in a Research Priority Report. This report outlines the top three comparative clinical effectiveness research (CER) priorities identified by the participants during the online community conversation.

Activities:

We hosted a four-hour online community conversation through the secure Genetic Support Foundation (GSF) Zoom platform to identify patient-centered CER topics. The session engaged an Advocacy Team of 24 participants, with balanced representation of both people with achondroplasia and parents of children with achondroplasia. Participants represented a diverse range of racial, ethnic, socioeconomic, geographic, and age backgrounds (see Table 1). Participants were recruited through established community networks and email outreach distributed by LPA.

Recruitment and engagement efforts were led by Andrea Schelhaas, MS, CGC. Ms. Schelhaas is a lifetime member of LPA, serves on the LPA Biotech Industry Liaison Committee and Medical Advisory Board, and has raised her children within the dwarfism community. To ensure balanced representation, inclusion, accountability, engagement, and sustainability, the project also provided equitable compensation to all participants ($125/hour), offered plain language materials and bilingual interpretation, conducted end-of-session evaluations, and included LPA representatives.

The Advocacy Team received PCORI Research Fundamentals Training led by Dr. Stephanie Meredith, a disability and health expert who has led two previous PCORI engagement projects. The session utilized a Community Conversation model in which participants engaged in facilitated dialogue rooted in participatory action research (PAR), with the goal of fostering collaboration and shared learning.[16,17]

Small group discussions were facilitated by community members serving as table hosts. Through this structured format, participants engaged in three separate breakout sessions, each followed by a full-group report-out. GSF staff members, including genetic counselors Andrea Schelhaas, Katie Stoll, and Katia Vine, along with Dr. Meredith and Mariah Chavez (a LEND graduate student), were distributed across breakout groups to document key findings and provide support as needed. Spanish translation was provided by Katia Vine, who is also a certified medical interpreter.

Guiding questions were intentionally broad at the outset to elicit a wide range of perspectives and then progressively narrowed to focus on patient-centered CER questions.

1. Concerns

What concerns do you feel are most important to individuals with achondroplasia and parents of children with achondroplasia?

2. Research Topics

Based on the concerns identified, what research topics do you think are most important to the achondroplasia community?

3. Patient-Centered CER Questions

How could these topics be turned into patient-centered CER questions? (What questions could research answer to compare different approaches to care and improve outcomes?)

Project Participants

Recruitment efforts yielded 74 responses. International respondents, duplicate registrants, and members of the same household were excluded. One participant withdrew due to a scheduling conflict. Participants and alternates were then selected to ensure a diverse range of perspectives. Demographics of the 24 individuals who participated are outlined below.

Table 1: Participant Characteristics

Characteristic	n=24 (%)
Role	
Parents of child with achondroplasia	12 (50.0%)
Adults with achondroplasia	8 (33.3%)
Dual Role (Adult with achondroplasia & Parent)	4 (16.7%)
Gender	
Women	16 (66.6%)
Men	8 (33.3%)
Age (Participant)	
18-25	2 (8.3%)
26-35	7 (29.2%)
36-45	7 (29.2%)
46-55	6 (25%)
56-65	1 (4.2%)
66-75	1 (4.2%)
Racial Identity	
White	18 (75%)
Black/African American	1 (4.2%)
Asian	1 (4.2%)
Multiracial (Asian/White)	1 (4.2%)
Self-describe (Latinx)	1 (4.2%)
Prefer not to answer	2 (8.3%)
Ethnicity	
Hispanic or Latine	3 (12.5%)
Not Hispanic or Latine	21 (87.5%)
Region [a]	
Northeast (CT, MA, ME, NH, NJ, NY, PA, RI, VT)	3 (12.5%)
Midwest (IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, SD, WI)	6 (25.0%)
South (AL, AR, DC, DE, FL, GA, KY, LA, MD, MS, NC, OK, SC, TN, TX, VA, WV)	7 (29.2%)
West (AK, AZ, CA, CO, HI, ID, MT, NM, NV, OR, UT, WA, WY)	8 (33.3%)

[a] Geographic regions are defined by the US Census Bureau

Table 2: Pediatric Data and Clinical Trial Engagement

Note: Treatment engagement data included only participants with children age 18 or younger (n=14) to reflect the relatively recent FDA approval of pharmaceutical therapies to increase height.

Category	
Age of Participant's Child/Children (Total)	**n=16 (%)**
Under 5	4 (25.0%)
5-9	2 (12.5%)
10-14	6 (37.5%)
15-18	2 (12.5%)
Over 18	2 (12.5%)
Clinical Trial Participation	**n=14 (%)**
Yes	2 (14.3%)
No	12 (85.7%)
Medication Use	**n=14 (%)**
Yes	6 (42.9%)
No	8 (57.1%)

Key Observation:

Decision-making varied significantly by parental role. Among parents without achondroplasia, 50% (n=6) reported pursuing a pharmaceutical therapy for their child. In contrast, none of the parents with achondroplasia pursued a pharmaceutical therapy or clinical trial to increase height for their child.

Findings: Notes from March 30, 2026 Online Community Conversation

Question 1: What concerns do you feel are most important to individuals with achondroplasia and parents of children with achondroplasia?

Group 1: Accessibility, Inclusion, and Identity/ Confidence-Building

- Limited access to adaptive products, which are often difficult to find and are expensive.
- Need for increased education in school settings about achondroplasia, including promoting respect, understanding boundaries, and fostering inclusion.
- Importance of developing confidence so that individuals with achondroplasia are not negatively impacted by comments or pushed toward medication use.

Group 2: Diagnosis Experience, Representation, Inclusion, and Identity/ Confidence-Building

- At time of diagnosis, families are often primarily educated about medical concerns by a provider or individual without achondroplasia, rather than about the robust and meaningful lives of people with achondroplasia. Participants were concerned about messaging that frames achondroplasia as something that needs to be fixed, particularly when families seek information online. There is a need for more balanced, strengths-based information and connection with the achondroplasia community at the time of diagnosis.
- Media representation impacts public perceptions of people with achondroplasia, highlighting the need for broader and more accurate representation.
- Concerns about how disability is perceived and discussed within the achondroplasia community, including the need to reduce stigma and support inclusion within the broader disability community.

Group 3: Emotional Well-Being/Mental Health, Care Navigation, Support Systems, and Diagnosis Experience

- Mental health and social concerns, including feelings of not fitting in, experiences of discrimination, and incidences of bullying in childhood and adulthood. The importance of helping children and others with achondroplasia develop confidence and tools to respond to discrimination.
- Limited access to specialized care, particularly in small towns or rural communities, often requiring travel to clinicians with expertise in achondroplasia.
- Need for support at the time of diagnosis to help families navigate doubts or questions, the lack of centralized resources addressing both health care and lived experiences, and the importance of guidance throughout the process.

Group 4: Provider Communication and Emotional Well-Being/Mental Health

- Need for mental health resources and improved provider access to information about achondroplasia.
- Importance of providers being open to feedback from individuals with achondroplasia and using intentional language.
- Importance of providers addressing the social and emotional needs of the community.

Question 2: Based on the concerns identified, what research topics do you think are most important to the achondroplasia community?

Group 1:

- **To address the lack of affordable adaptive resources**: Analyze the impact of disability benefits administration policies (such as CA SB 1090), corporate–public partnerships (such as accessibility initiatives at Sonic or airports), public policies (such as the ADA), website compiling lists of adaptations and accommodations, and training programs related to adaptive accommodations. Assess how these interventions impact the availability of adaptive resources, accessibility, and quality of life for people with achondroplasia.
- **To address the impact of potentially biased marketing materials from pharmaceutical companies**: Examine how online information affects the confidence of people with dwarfism. Identify ways to improve access to balanced resources related to self-confidence and self-advocacy that are independent of pharmaceutical or surgical interventions.
- **To address the limited understanding of achondroplasia among new parents**: Evaluate the impact of peer mentorship and resources about local chapters.

Group 2:

- **To address limited provider knowledge of achondroplasia**: Assess current sources of provider-facing information about achondroplasia. Identify key knowledge gaps and/or areas of misunderstanding.

- **To address barriers to community connection**: Assess the impact of structured mentorship programs for new parents of children with achondroplasia, such as peer mentorship models similar to Big Brothers Big Sisters.

- **To address disability stigma:** Evaluate how people with achondroplasia understand and relate to the broader disability community. Assess the impacts of education that addresses disability identity affirmation, disability rights awareness, governmental supports, legal protections, and access to community tools and resources.

Group 3:

- **To address limited availability of resources**: Assess the potential impact of providing local resources to new parents and developing a registry of knowledgeable specialists connected to the achondroplasia community.
- **To address limited peer connection opportunities for older children with achondroplasia**: Evaluate the potential impact of expanding mentorship programs, such as Big Brothers Big Sisters, and facilitated parent-to-parent connections.
- **To address limited access to specialists**: Examine the potential health benefits of mobile specialty clinics.
- **To address lack of community integration and inclusion**: Assess the impact of community outreach and exposure initiatives, such as an LPA Lending Library.

Group 4:

- **To address mental health concerns**: Evaluate the impact of mentorship and mental health supports on the mental health and social well-being of teens with intersectional identities.
- **To address access to knowledgeable care**: Assess access to medical professionals with adequate education on achondroplasia. Evaluate how receiving balanced, accurate information at the time of diagnosis impacts health outcomes.
- **To address early treatment decision-making**: Examine parental motivations for opting into treatment at ages when their child cannot provide input, how communication of updated and accurate information shapes parental treatment decision-making, and the impact of treatment options on children's mental health.

Question 3: How could these topics be turned into patient-centered CER questions? (What questions could research answer to compare different approaches to care and improve outcomes?)

Group 1

- For average-height parents of children with achondroplasia, how do early referrals to advocacy organizations with parent mentor programs and resources, compared with medical, pharmaceutical, or internet-based information alone, affect parent mental health, decision-making, and child identity formation?
- For people with achondroplasia, how do current ADA laws, public and corporate spaces, and adaptive resource guides affect awareness of available accommodations and self-advocacy skills and meet community accessibility needs?

Group 2

- How do formal resources (e.g., educational materials, clinical guidance, and toolkits) compare with peer mentorship (e.g., connections with others with achondroplasia) in helping individuals and families understand lived experiences and support positive identity development?
- Among new or expectant parents of children with achondroplasia, how does connection to adults with achondroplasia (e.g., through peer mentorship), compared with standard educational resources alone, impact parent understanding of the diagnosis, emotional adjustment, and support for positive identity development in their child?
- How do public education campaigns about disability, compared with traditional educational awareness materials, influence understanding, attitudes toward disability, and inclusion?

Group 3

- Are people with achondroplasia more likely to receive guideline-based standards of care in mobile clinics compared with training local providers?
- For children with achondroplasia, how do formal mentorship programs (such as the Big Brothers Big Sisters) compare with parent-established connections outside of LPA in supporting social and emotional well-being?

Group 4

- How does provider use of an affirmative language tool developed with guidance from individuals and parents with achondroplasia impact children's perspectives of their diagnosis and care decisions, compared with care provided without use of affirmative language tools?
- How does acceptance within the family unit, larger community, or other support systems impact self-perception among individuals with achondroplasia participating in mentorship programs compared with those without access to a structured support system?
- Among individuals using Voxzogo and similar drugs, compared with children who do not use these medications, are there differences in the frequency of common health complications such as spinal stenosis, sleep apnea, or the need for ear tubes?

Additional thematic analysis of identified concerns and research topics can be found in the Appendix.

Top 3 Research Priorities and Patient-Centered Comparative Clinical Effectiveness Research Questions Identified

After reviewing the proposed research priorities and patient-centered comparative clinical effectiveness research (CER) questions, several key overlapping themes emerged. These themes were synthesized into the following top three CER areas. Priorities were selected based on frequency of discussion, relevance across multiple domains (e.g., medical, educational, social/emotional, etc.), and potential for measurable outcomes.

Priority 1: Address societal discrimination and mental health needs of people with achondroplasia

Examine how stigma, bias, access to accommodations, and competing narratives about disability and treatment influence identity development, confidence, and/or resilience, particularly among individuals with intersectional identities.

Identified Areas of Interests Related to the Above Include:

- Marketing and communication of height-altering interventions (pharmaceutical and surgical)
- Structured peer mentorship programs
- Community perceptions of disability identity and stigma
- Community outreach and education initiatives
- Public media representation and perceptions
- Access to culturally responsive mental health supports

CER Question: Among young adults with achondroplasia, particularly those with intersectional identities, how do social and identity-affirming supports, compared with approaches focused primarily on height-altering interventions, affect confidence, identity development, community inclusion and participation, and/or experiences of stigma and discrimination? (Groups 1, 2, 3, & 4)

Related Concerns Addressed:

- Supporting identity development, social and emotional well-being, and confidence (Groups 1, 2, 3, & 4)
- Addressing medical and societal discrimination, including stigma and media representation, and assessing their impacts on mental health (Groups 1, 2, 3, & 4)

Priority 2: Improve access to medical care and accommodations

Address gaps in healthcare access and adaptive supports within traditional care delivery models. Examine how barriers to access affect health outcomes, community participation, and/or quality of life.

Identified Areas of Interests Related to the Above Include:

- Public policy and systems-level barriers
- Centralized resources and training repositories related to accommodations and adaptations
- Gaps in care delivery models
- Innovative care delivery models (e.g., mobile care clinics, telehealth, etc.)

CER Question: For individuals with achondroplasia and their families, how do enhanced care delivery approaches (e.g., mobile care clinics, telehealth, and access to adaptive equipment), compared with usual care, affect access to health services, health outcomes, and/or quality of life? (Groups 1, 3, & 4)

Related Concerns Addressed:

- Access to adaptive equipment and products (Group 1)
- Access to specialized health care (Group 3)
- Public policy and systems-level barriers (Group 1)
- Gaps in care delivery models, particularly in rural areas, and the potential impact of innovative care models (Group 3)

Priority 3: Support new parents and early treatment decision-making

Improve access to balanced, affirming support resources for new and expectant parents to promote informed decision-making, parent well-being, and/or positive identity development in children with achondroplasia.

Identified Areas of Interests Related to the Above Include:

- Peer-parent mentorship
- Connections to local advocacy communities
- Clinician training in balanced, affirming communication
- Development and dissemination of balanced educational resources informed by lived experience
- Creation of a more comprehensive registry of providers experienced in achondroplasia care

CER Question: CER Question: Among new and expectant parents of children with achondroplasia, how do affirming care models, compared with usual medically-centered care, affect parent understanding and preparedness, parent and child well-being, medical decision-making, and/or positive identity development? (Groups 1, 2, 3, & 4)

Related Concerns Addressed:

- New parents receiving information about achondroplasia that focuses primarily on medical concerns (Group 1, 2, 3, & 4)
- Concerns related to stigma and discrimination in how the diagnosis is communicated (Group 1, 2, 3, & 4)
- Improving and access to support and understanding of lived experience at the time of diagnosis to foster informed decision-making; including through strategies such as peer-parent mentorship, parent educational resources, and/or connections to local advocacy communities (Groups 2 & 3)

Future Directions

The online community conversation is one part of a three-part project. Future project activities include a two-hour breakout session at the Little People of America (LPA) national conference in New Orleans, LA, in July 2026. This session will bring together individuals with achondroplasia, family members, providers, and researchers.

The goals of this conversation are to 1) further refine the patient-centered CER priorities identified through **Aim 1**, 2) identify effective and inclusive strategies for engaging the broader achondroplasia community in research, and 3) document findings in a Research Brief (**Aim 2**). This brief will outline a strategic plan for advancing patient-centered CER through multi-stakeholder engagement and partnerships that reflect the needs and priorities of the achondroplasia community.

Aim 3 focuses on disseminating the identified research priorities and engagement strategies and supporting their integration into future research initiatives. This will include a follow-up convening with a Professional Team of achondroplasia providers, researchers, pharmaceutical representatives, and advocacy leaders.

The goal of Aim 3 is to foster multidisciplinary collaboration and sustained partnerships to advance future patient-centered CER. Insights from Aim 3 will be incorporated in the final Research Brief. In collaboration with LPA, project staff will disseminate the final Research Brief to community members, advocates, clinicians, and researchers, with the goal of elevating patient and community voices in future achondroplasia research initiatives.

Evaluation Results

23 of 24 (96%) of participants completed a post-meeting evaluation.

- All respondents (100%) agreed or strongly agreed that:
  - The community conversation accomplished the goal of identifying the top patient-centered research priorities for people with achondroplasia and parents of children with achondroplasia.
  - The event produced strategies and recommendations that will help to improve the PCOR (patient-centered outcomes research) involving people with achondroplasia, if implemented.
  - The conversation was a valuable use of their time.
  - They would invite someone they knew to attend another event like this if one in the future.
- Most respondents (91%) agreed or strongly agreed that their input was valued. One adult with achondroplasia and one parent of a child with achondroplasia (9%) neither agreed nor disagreed that their input was valued.

Participant Quotes

Selected participant quotes from the evaluations are included to further illustrate these findings.

"Thank you very much for this opportunity. I don't often have the chance to connect with people with ACHON and/or parents, so hearing their perspectives and experiences was very educational for me. It made me feel heard and part of a community I'm not used to being part of. I look forward to seeing these great ideas become part of our medical system."
– parent of child with achondroplasia

"I really enjoyed these conversations! I only wish that we had more time!"
– adult with achondroplasia

"This was wonderful. I wish we had more time! Many friends and I with dwarfism have been hoping that our voices could be heard beyond a piece of medical paper (or a provider who hasn't even met someone like us), so thank you so much for this opportunity."
– adult with achondroplasia who is also a parent of child with achondroplasia

"Very thoughtful discussion and setup. Appreciated the intention behind it all and the importance of topics that are not normally studied."
– parent of child with achondroplasia

"Amazing experience to be a part of and I am so glad to have joined the conversation!"
– adult with achondroplasia

"It's great to hear common problems from people with different backgrounds!"
– parent of child with achondroplasia

"This was a great opportunity to express my own concerns and ideas. It was a little tricky coming up with the CER topics, but I loved the conversation we had. Thank you!"
- parent of a child with achondroplasia

Report Review

Participants were given the opportunity to review a draft of the Research Priority Report prior to publication. Fifteen of 24 participants (62.5%) completed a review using a custom rubric to help ensure the report accurately reflects the community conversations and perspectives shared throughout the process. Participant feedback was incorporated into the final draft.

Appendix: Concerns and Research Topics Coded by Theme

Concerns: *Participants were asked to identify the most important concerns for individuals with achondroplasia and parents of children with achondroplasia. Responses reflected a range of experiences and were organized into key domains including: health care access and coordination, treatment decision making, mental health, stigma, and social inclusion, family dynamics, community-informed resource development, and mentorship and empowerment.*

Domain 1: Healthcare Access, Quality, and Coordination
Barriers to accessing knowledgeable, coordinated, and affirming healthcare were outlined. Participants emphasized the need for systems that are responsive to medical concerns and reflective of lived experience.

Key Themes:

- **Access to Specialized Care:** Travel burden and difficulty identifying experienced providers (1, 3, 4)
- **Insurance and Structural Barriers:** Disruptions due to network changes; need for expanded Medicaid access (3, 4)
- **Care Coordination Needs:** Parent burden of staying on top of monitoring for common medical concerns; benefits of patient navigators and integrated care models (1, 3)
- **Provider Knowledge and Preparedness:** Limited provider knowledge, reluctance to seek robust education/information about the diagnosis, delayed diagnosis, lack of understanding of achondroplasia-specific growth charts/milestones, and setting unrealistic or potentially harmful expectations (1–4)
- **Burden on Families:** Patients and caregivers often act as experts or educators for providers (1, 3)
- **Communication Needs and Barriers:** Importance of intentional language and validation of lived experience (2–4)

Domain 2: Treatment Decision-Making and Healthcare Outcomes
Participants described the need for balanced, patient-centered approaches to treatment discussions.

Key Themes:

- **Perspectives on Interventions:** Concerns about intent and framing of height-altering interventions including pharmaceutical interventions and limb lengthening (3, 4)
- **Patient-Centered Care:** Emphasis on support beyond a medical model intended to "fix"; recognition that individuals are more than their diagnosis (1, 2, 4)
- **Role of Environmental Supports:** Accessibility and accommodations as critical components of well-being (1)

Domain 3: Mental Health, Stigma, and Social Inclusion
Social stigma, discrimination, and representation were highlighted as impacting mental health and quality of life across the lifespan

Key Themes:

- **Bullying and Discrimination:** Concerns in community, school, and workplace settings (3, 4)
- **Social Acceptance:** Universal need for improved inclusion and social acceptance
- **Emotional Support Needs:** Access to culturally-appropriate mental health resources for individuals and families
- **Representation and Language:** Harmful public discourse including use of the "M-word," mascots and negative media portrayals; emotional burden of continued advocacy (2, 3)
- **Information Integrity:** Concerns about inaccurate or biased messaging from external stakeholders including pharmaceutical companies (1, 2)

Domain 4: Family Dynamics and Diversity in Lived Experience
Experiences vary based on role and family and community support systems.

Key Themes:

- **Diverse Perspectives:** Differences among adults with achondroplasia, parents with the condition, and average-height parents (4)
- **Family Dynamics:** Variability in understanding and acceptance within families
- **Parenting Experience:** Concerns about parenting a child with an unfamiliar diagnosis (4)

Domain 5: Information Access, Resources, and Community Integration
Participants highlighted gaps in access to clear, centralized, and community-informed information and resources.

Key Themes:

- **Diagnosis Delivery:** Importance of inclusive, strengths-based materials and communication at diagnosis (1)
- **Information Gaps:** Limited information guided by lived-experience and lack of centralized resources (2)
- **Medical Information Needs:** Desire for accessible, up-to-date, and practical resources including resources for medical appointments (4)
- **Community Connection:** Need for clearer understanding of available supports within advocacy organizations and improved integration with broader disability communities (2, 4)
- **Adaptive Tools:** Limited awareness of and access to adaptive products and and environmental modifications (1)
- **Equity in Access:** Importance of information that is accessible to people from varied education backgrounds and the need for resources that are culturally and linguistically appropriate. Participants also highlighted concerns with availability of interpretation services and variability of resources by state/region (1,4)

Domain 6: Mentorship, Advocacy, and Empowerment
The importance of mentorship, advocacy development, and community engagement were outlined as a means to empowering individuals and families.

Key Themes:

- **Mentorship Opportunities:** Strong interest in mentorship across a variety of formats, particularly from individuals with achondroplasia to parents who are unfamiliar with the condition (1–4)
- **Self-Advocacy Skills:** Importance of building advocacy skills and confidence from an early age (1)
- **Community Education:** Opportunities for outreach and awareness including in schools and within the community (1, 3)

Research Topics:

Healthcare Access and Quality of Care

- Improved dissemination of accurate, up-to-date medical information on achondroplasia and evaluation of where clinicians currently access information (2)
- Opportunities to establish a centralized, trusted source for clinical guidance (2)
- Impact on care when working with providers who lack knowledge of achondroplasia (1)
- Impact on health outcomes when families receive accurate information at diagnosis and/or access to experienced medical professionals (1, 4)
- Development of a "patient experience" provider registry or community-informed database to identify experienced clinicians (3)
- Use of tools (e.g., QR codes) to connect families to trusted resources and guidelines at diagnosis (3)
- Exploration of decentralized and mobile specialty care models to bridge geographic gaps (3)

Clinical Care and Care Navigation, Treatment Outcomes, and Decision-Making

- Communication about and access to current research on height-enhancing therapies and outcomes (1, 4)
- Inclusion of diverse perspectives on height-enhancing treatments (1, 2)
- Treatment approaches for spinal stenosis (4)
- Creating supportive environments for complex medical conversations (4)
    - Use of tools (e.g., questionnaires) to support youth engagement in care discussions (4)
- Parent intentions and considerations when opting into early treatment (i.e. when children are too young to provide input about treatment decisions) (1, 4)
- Impact of early treatment decision-making on children's autonomy and mental health (1, 4)

Mental Health, Stigma, and Identity

- Impact of affirming language in clinical settings on social-emotional outcomes (1, 4)
- Effects of messaging to parents on child self-confidence and family communication (1)
- Strategies to support and improve mental health and overall well-being (2)
- Tools to help individuals and families respond to discrimination (2)
- Impact of access to mental health supports in (4):

    - The general sense
    - During treatment discussions
    - Research enrollment
- Influence of family acceptance on care experiences and outcomes (4)
- Effects of intersectional identities on mental health and identity development (4)
- Influence of media representation on perceptions and self-concept (1)

Community Resources and Support Systems

- Implementation and evaluation of different mentorship models (e.g. peer mentors and "Big/Little" programs) on: (1, 2, 3, 4)
    - Decision-making confidence
    - Identity development
    - Mental health
- Addressing isolation between community events and strengthening ongoing support networks (3, 4)
- Development of resources that facilitate access to lived experience and peer mentorship (2)
- Role of advocacy community connections (e.g., LPA) in shaping outcomes and support (1, 4)
- Evaluating best practices for parent groups to support new parents of children with skeletal dysplasia as determined by a convening group (1)

Accessibility and Policy

- Research on accessibility gaps and adaptations (1)
- Research on ADA-related policies as they apply to individuals with achondroplasia
- Impact of disability benefits, partnerships, and accommodation programs on accessibility and quality of life

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