Improving guideline-directed management of achondroplasia: results of a pre-implementation study

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Introduction

- Achondroplasia is the most common form of skeletal dysplasia and may lead to multisystem complications that require lifelong management[1–3]
- The adoption of evidence-based achondroplasia treatment guidelines into clinical practice is variable, and individuals with achondroplasia may not consistently receive the full extent of recommended care[4,5]
- Implementation science identifies strategies to accelerate the adoption of evidence-based guideline-directed therapy and improve patient care[1,6-9]
- By leveraging large-scale real-world data, implementation science can identify gaps between recommended care and clinical practice and develop strategies to close these gaps

Objectives

- Present the design of an implementation science study that aims to identify strategies for closing gaps in achondroplasia care
- Present preliminary results for guideline synthesis and the identification of high-priority statements for quality assessment and improvement

Methods

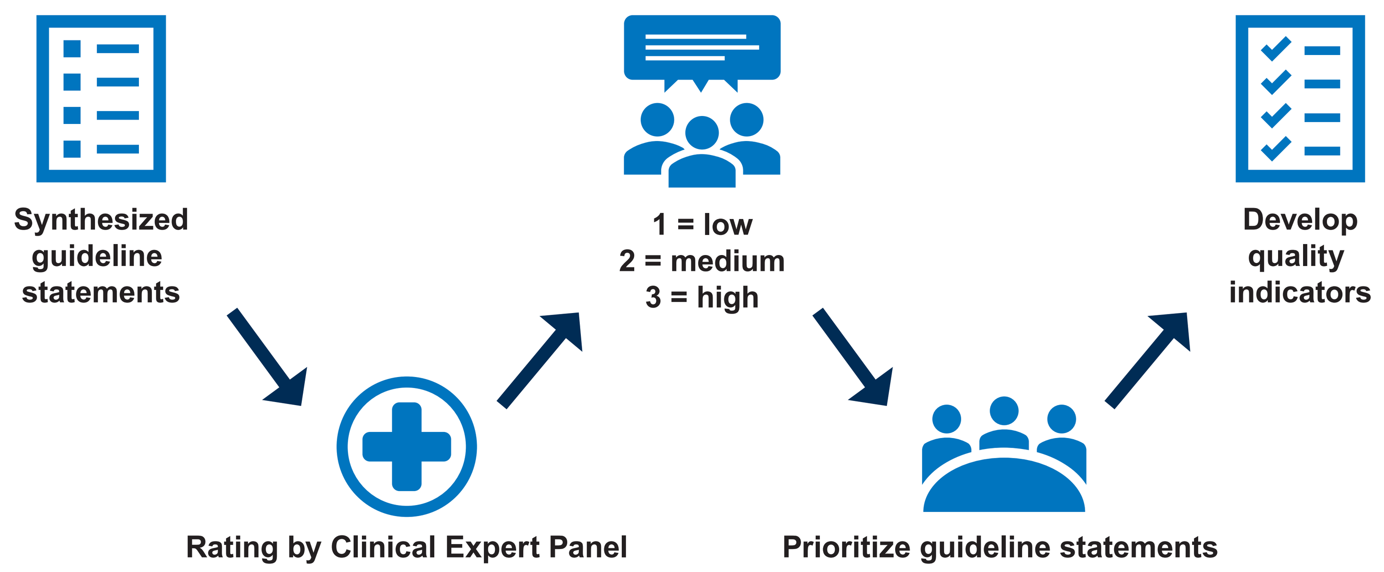
Study protocol

1 Measure achondroplasia prevalence

- Generate estimated case counts in Kaiser Permanente Southern California and other regions

2 Prioritize guideline recommendations

- Extract individual guideline recommendation statements from the 3 primary achondroplasia clinical practice guidelines relevant for US health care delivery[1,8,9]
- Incorporate recommendations (N = 389) in a structured rating form for independent rating by 4 clinical experts (2 MDs, genetic counselor with lived experience, PharmD with lived experience)
- Raters assessed (1) magnitude of care gap, (2) importance of adherence for patient outcomes, and (3) feasibility of improvement; total rating score indicates each recommendation's level of priority for improvement (high, medium, low)



3 Quality indicator development: multi-stakeholder perspectives

- Convert guideline statements rated high priority to quality indicators following SMART criteria (Specific, Measurable, Achievable, Relevant, and Timely)
- Validate and augment guideline-derived quality indicators with perspectives from individuals with lived experience
- Refine the draft guideline-based and lived experience-based quality indicators through review and consensus among the Clinical Expert Panel

4 Design implementation strategy

- Propose a generalizable implementation strategy bundle to guide localized design and adaptation of implementation strategies to close quality gaps

Results

- Recommendation statements were categorized as age band-specific or specific to various types of specialty care; the highest priority recommendations address care for children and treatment with vosoritide
- **Table 1** lists the highest priority recommendations based on the average total score across all raters; total score (range 3–9) is the sum of individual scores for (1) magnitude of care gap, (2) importance of adherence for patient outcomes, and (3) feasibility of improvement (each rated 1 = low, 2 = medium, 3 = high)

Table 1. Recommendations rated highest priority based on preliminary findings

Recommendation category	Overall rating	Care gap	Importance	Feasibility of improvement
Age bands				
Assess growth and development in comparison only with children with achondroplasia	8	2	3	3
Because most children with achondroplasia snore, monitor closely for signs of obstructive sleep apnea. If obstructive sleep apnea is suspected, then pulmonary consultation and polysomnography are indicated	8	2.5	3	2.5
Specialty areas				
Patients should be monitored for adverse effects at all follow-up appointments	8	2.5	3	2.5
When annual height velocity has slowed to <1.5 cm per year, radiography should be performed to check the status of the growth plates; if they are closed, treatment with vosoritide should be stopped	8	2.5	3	2.5

Age bands include 3 broad categories: “prenatal, childbirth, and immediate postnatal periods,” “childhood,” and “adolescence and adulthood.” Specialty areas include 5 broad categories: “spine and genu varum,” “limb lengthening, vosoritide,” “respiratory, sleep; ear, nose, and throat,” “orthodontics and maxillofacial surgery; anesthesia,” and “pain and function; nutrition, physical activity, and weight management.”
Overall rating is the sum of individual mean scores for (1) magnitude of care gap, (2) importance of adherence for patient outcomes, and (3) feasibility of improvement (each rated 1 = low, 2 = medium, 3 = high). Other data represent the mean score for each rating dimension.

- Raters showed significant disagreement in their preliminary ratings for 2 recommendations (**Table 2**)
  - Disagreement reflected differing opinions regarding the availability of staff and other resources required to adhere to the recommendations
- Disagreement in preliminary ratings for magnitude of care gap was high for 1 recommendation, indicating varying opinions regarding patterns of care across the US

Table 2. Recommendations showing the highest level of disagreement

Recommendation category	Overall rating	Care gap	Importance	Feasibility of improvement
Age bands				
Adolescents with achondroplasia might benefit from the support of dedicated professionals to assist with adjustment to their condition and the development of coping strategies for school, employment, and social environments	6 (4–8)	1.5 (1–2)	2.5 (2–3)	2 (1–3)
Specialty areas				
Individuals with achondroplasia are at risk of respiratory failure during respiratory tract infections, justifying preventative measures such as the avoidance of passive smoking and contact with infected persons. In these patients, respiratory syncytial virus prophylaxis with monoclonal antibodies can be considered	6 (4–8)	2 (1–3)	2.5 (2–3)	1.5 (1–2)
In patients >5 years of age, assessments should include the STEMS and the ASK	6 (4–8)	1.5 (1–2)	2.5 (1–2)	2 (1–3)

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Overall rating (range, 3–9) is the sum of individual mean scores for (1) magnitude of care gap, (2) importance of adherence for patient outcomes, and (3) feasibility of improvement (each rated 1 = low, 2 = medium, 3 = high). Other data represent the mean score (range) for each rating dimension.
ASK, Activities Scale for Kids; STEMS, Screening Tool for Everyday Mobility and Symptoms.

- Preliminary ratings revealed several recommendations rated as “high” for magnitude of care gaps and importance of adherence for patient outcomes, but low feasibility of improvement (**Table 3**)
- In each case, feasibility of improvement was rated low due to limitations in access to specialized, experienced care, particularly in rural and suburban areas for which tertiary and quaternary care centers were not present and patient volume was insufficient to support specialized achondroplasia and skeletal dysplasia teams

Table 3. Recommendations showing significant discrepancies between need for improvement and feasibility of improvement

Recommendation category	Overall rating	Care gap	Importance	Feasibility of improvement
Age bands				
All children with achondroplasia should receive regular follow-up by a multidisciplinary team, guided by a health-care professional with expertise in achondroplasia	6.5	2.5	3	1
Pregnant women with achondroplasia should undergo an antenatal anesthetic clinical assessment	6	2.5	2.5	1
In asymptomatic infants with achondroplasia, MRI scanning to evaluate the cervicomedullary junction and foramen magnum size should be considered during the first months of life	7	2.5	3	1.5
Foramen magnum decompression should be carried out by a neurosurgical team with prior experience in performing this procedure	7	2.5	3	1.5
Specialty areas				
Surgical procedures, such as osteotomies or guided growth to correct lower limb malalignment, should be performed in centers of excellence in achondroplasia	7	2.5	3	1.5
Patients with achondroplasia, especially children, should be anesthetized preferentially in hospitals where care providers are knowledgeable and experienced in caring for patients with achondroplasia and skeletal dysplasias	7	2.5	3	1.5
Due to foramen magnum stenosis, head and neck movement should be avoided or minimized during bag-mask ventilation and intubation of patients with achondroplasia	7	2.5	3	1.5
Chronic pain should be considered as a multifactorial problem and is best managed in individuals with achondroplasia through a multidisciplinary approach	6	2	3	1

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Overall rating is the sum of individual mean scores for (1) magnitude of care gap, (2) importance of adherence for patient outcomes, and (3) feasibility of improvement (each rated 1 = low, 2 = medium, 3 = high). Other data represent the mean score for each rating dimension.
MRI, magnetic resonance imaging.

Conclusions

- Implementation science can be applied to rare disease domains like skeletal dysplasias to facilitate the integration of evidence-based guideline-directed therapy into routine clinical practice
- This study presents a preliminary categorization of guideline recommendations for achondroplasia based on importance of adherence, room for improvement, and feasibility for improvement
- These preliminary results demonstrate the feasibility of prioritizing guideline recommendations for future quality implementation efforts to improve the diagnosis, treatment, and care of people with achondroplasia

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