

Medical and Surgical Concerns in Paediatric Patients with Achondroplasia: A Retrospective Chart Review

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ABSTRACT

Background: Achondroplasia is the most common skeletal dysplasia and is associated with a wide range of medical and surgical comorbidities beyond short stature. Comprehensive descriptions of the clinical manifestations and management of these comorbidities within paediatric populations remain limited. We aimed to describe the spectrum of medical and surgical comorbidities and their management in paediatric patients with achondroplasia seen at a tertiary care centre.

Methods: We created a registry of patients seen at the Skeletal Dysplasia Clinic and conducted a retrospective cohort study of 60 patients with achondroplasia seen between January 2018 and October 2021. We collected longitudinal data on referrals, medical concerns, and medical and surgical management.

Results: Among the 60 patients included, neurological and orthopedic concerns were commonly observed. Foramen magnum stenosis/cervicomedullary compression required surgery in 37% of patients. Genu varum was treated operatively in 23% of patients. Central sleep apnea affected 25% of patients, and obstructive sleep apnea affected 62%. Fifty-three percent of patients underwent tonsillectomy/adenoidectomy surgery, and 18% received a continuous positive airway pressure (CPAP) machine. Auditory concerns were also prevalent, and myringotomy tubes were inserted in 48% of patients.

Conclusion: Paediatric patients with achondroplasia experience a broad range of medical and surgical comorbidities requiring multidisciplinary management. This study describes the spectrum of clinical manifestations and management strategies observed in a tertiary care setting and highlights the complexity of care required for this population.



KEYWORDS

Achondroplasia, Pediatrics, Disease Management

1 | INTRODUCTION

Achondroplasia is the most common skeletal dysplasia, with an estimated incidence at birth of 1 in 25,000 to 1 in 30,000. (1) Achondroplasia is typically caused by a de novo, autosomal dominant variant in the fibroblast growth factor receptor type 3 (FGFR3) gene with a glycine to arginine substitution at codon 380 (G380R), although rare exceptions to this variant occur. (1–4) The substitution in FGFR3 activates inhibitory processes that downregulate chondrocytic bone growth, (1,5) resulting in dwarfism and potential orthopedic, neurosurgical, and respiratory comorbidities. (6)

Foramen magnum stenosis in achondroplasia can result in neurologic problems (e.g., central sleep apnea) and pain, often necessitating surgical decompression. Orthopedic concerns due to metaphyseal irregularities and joint laxity may lead to orthopaedic concerns such as genu varum and pain necessitating surgical intervention. Midface hypoplasia, enlarged adenoids/tonsils, and short eustachian tubes may predispose children to respiratory infections, otitis media, and hearing loss. (7) The comorbidities associated with achondroplasia may adversely impact children's health and quality of life. (8) Management typically requires lifelong involvement from multidisciplinary healthcare providers.

The Hospital for Sick Children (SickKids) is a leading tertiary paediatric care and research centre in Canada, providing specialized care for children with rare and complex conditions (9). It cares for children with achondroplasia, including routine follow-up, treatments, and surgeries. Given the heterogeneous population and complex medical needs of children with achondroplasia seen at SickKids, it is essential to systematically track their symptoms and medical intervention for clinical teams to have up-to-date information about the patient's clinical status and medical management over time. We created a site-specific registry to summarize the characteristics of children with skeletal dysplasia at SickKids to track this population's characteristics and medical and surgical management at Canada's largest children's hospitals.

1.1 | Aims

We conducted a retrospective cohort study to describe and characterize the spectrum of medical and surgical comorbidities and their management in paediatric patients with achondroplasia seen at a tertiary care centre. Specifically, the objectives of this study were to (a) describe the frequency and types of abnormalities identified through recommended achondroplasia screening and clinical assessments, and (b) summarize the medical and surgical management strategies employed for these comorbidities.

2 | METHODS

This study was approved by the Research Ethics Board (REB) at SickKids (REB# 1000059185). Given that this study used pre-existing patient data, we obtained a waiver of consent in line with institutional REB guidelines. Data were manually extracted from electronic medical records of patients seen at the Skeletal Dysplasia Clinic at SickKids between January 01, 2018, to October 31, 2021. Patient's medical records were examined for common medical concerns associated with achondroplasia, age of diagnosis or when concerns were first noted, medical and surgical management details, and effectiveness of management. Variables collected included demographic information (age, sex), genetic findings, neurological, orthopedic, respiratory, and auditory comorbidities identified through recommended screening, and associated medical and surgical management. To determine the efficacy of medical and surgical management, clinic notes were reviewed, and outcomes were categorized as resolved, partially resolved, or not resolved based on physician documentation and discussion with the research team. Data abstraction was performed by a single reviewer using a structured data collection framework developed by the research team based on study objectives and variables of interest. The framework included predefined fields for demographic characteristics, genetic findings, and clinical comorbidities (neurological, orthopedic, respiratory, and auditory), as well as associated medical and surgical management and outcomes. Management outcomes were recorded as documented in the electronic medical record by treat-

ing clinicians, without independent adjudication. For patients with multiple clinical concerns, outcomes were abstracted separately for each condition. Duration of symptoms and time to resolution were not consistently documented and therefore were not systematically analyzed. Body mass index (BMI) was calculated based on patients' most recent height and weight. Demographic variables collected included age and sex. Additional sociodemographic information was not consistently available in the medical records and was therefore not included in the analysis.

Data from patient charts were manually recorded into standard surveys on REDCap Internal (REDCap 12.3.3 - © 2022 Vanderbilt University). Data were processed using the Excel® software (Microsoft Corp. 2016) and analyzed using descriptive statistics. All percentages are reported as a percent of the total number of patients in the study (n = 60), unless stated otherwise. When reporting non-mutually exclusive subgroup indications, absolute numbers are presented without percentages to avoid misleading denominators.

3 | RESULTS

Sixty patients with achondroplasia were included in this study. The median age at the time of data collection was 10.1 years (range: 2.8-23.5 years). Thirty-one patients were male (52%) and 29 were female (48%).

Seven patients (12%) had inherited FGFR3 mutations. Fifty-two patients (87%) had a typical de novo mutation. One patient (2%) had an abnormal FGFR3 variant (de novo, c.835A>T, p.Ser279Cys). All patients had genetic testing. The most common comorbidities were foramen magnum stenosis, kyphosis, and recurrent otitis media. Findings are summarized in Table 1. Patients could have multiple comorbidities and therefore may be represented in more than one diagnostic category.

3.1 | Medical and Surgical Concerns and Management

3.1.1 | BMI and Gross Motor Development

Fourteen patients (24%) were considered overweight or obese. One patient in the obese category was seen by

dietetic nutrition for weight management. One additional obese patient was enrolled in an obesity management program.

The age at which the patients were first noted to sit independently and began walking was recorded as a guide for gross motor development. The age the child first started sitting was reported for 12 children. The median age was ten months (range: 8-15 months). The age when the child first started to walk was reported for 35 children. The median age was 20 months (range: 12-36 months).

3.1.2 | Neurological Concerns

Forty-five patients (75%) had foramen magnum stenosis, and 27 (45%) had cervicomedullary compression. All 22 patients (37%) who underwent cervicomedullary decompression surgery showed significant narrowing of the foramen magnum.

Following initial cervicomedullary decompression surgery, neurological concerns were fully resolved without the need for revision surgery in 15 patients. Symptoms were partially resolved in six patients who required revision surgery due to bony regrowth or recurrent or worsening symptoms. Among these six patients, symptoms were fully resolved after revision surgery in three patients. One patient experienced persistent symptom progression after the first revision but achieved full resolution following a second revision surgery. One patient had radiographic evidence of bony regrowth but remained asymptomatic and did not require further surgical intervention. One patient experienced chronic back and neck pain that did not resolve following revision surgery. Outcome data were not documented for one patient.

Seven patients underwent cervicomedullary decompression surgery for symptoms related to sleep apnea. Sleep apnea symptoms were fully resolved for three patients, partially resolved in one patient following two revision surgeries, and not resolved in three patients. For the patients whose sleep apnea symptoms did not resolve following surgical intervention, additional information regarding comorbidities or subsequent management was not available at the time of manuscript preparation.

Medical concern	Patient Count (percentage), n = 60	Median Age (min–max, years)
BMI		
Underweight (BMI < 18.5)	3 (5%)	6 (5–14)
Normal weight (BMI 18.5–25)	43 (72%)	8 (2–23)
Overweight (BMI 25–30)	7 (12%)	12 (5–18)
Obese (BMI > 30)	7 (12%)	17 (15–20)
Neurological complications		
Foramen magnum stenosis	45 (75%)	1.0 (0.0–16)
Cervicomedullary compression	27 (45%)	1.1 (0.3–11)
Cervicomedullary decompression surgery	22 (37%)	1.3 (0.4–12)
Cervicomedullary decompression revision surgery	6 (10%)	6.0 (0.5–18)
Second cervicomedullary decompression revision surgery	1 (2%)	2.2
Orthopedic complications: kyphosis		
Kyphosis Notes	38 (63%)	1.3 (0.8–11)*
Resolved spontaneously / No intervention required	37 (62%)	N/A
Intervention required	1 (2%)	11
Orthopedic complications: Genu Varum		
Genu varum requiring correction	14 (23%)	10 (6–17)
Guided growth	11 (18%)	10 (6–15)
Osteotomy	2 (3%)	16.5 (16–17)
Limb lengthening	1 (2%)	9
Respiratory complications		
Central sleep apnea	15 (25%)	1.1 (0.3–5)†
Obstructive sleep apnea	37 (62%)	1.6 (0.1–14)‡
Tonsillectomy/Adenoidectomy	32 (53%)	2.8 (0.8–9)§
Continuous positive airway pressure (CPAP)	11 (18%)	3.3 (0.7–13)
Auditory complications		
Recurrent otitis media	31 (52%)	2.0 (0.7–7)
Middle ear effusions	22 (37%)	1.6 (0.2–7)¶
Hearing loss	23 (38%)	3.0 (0.2–15)**
Myringotomy tubes	29 (48%)	2.3 (0.7–10)††

Note:

*Age of kyphosis was not specified for 10 patients.

†Age of CSA diagnosis was not specified for 1 patient.

‡Age of OSA diagnosis was not specified for 2 patients.

§Age of T/A was not specified for 1 patient.

||Age first diagnosed was not specified for 7 patients.

¶Age first diagnosed was not specified for 2 patients.

**Age first diagnosed was not specified for 1 patient.

††Age of first tube insertion was not specified for 2 patients.

TABLE 1 Summary of medical complications of achondroplasia patients

In addition to sleep apnea, one patient underwent cervicomedullary decompression surgery for pappilledema edema, urinary incontinence, sudden loss of consciousness and balance, and headaches, and two for myelopathy. All these concerns were fully resolved post-operatively. One patient experiencing severe developmental delays demonstrated no improvement post-operatively. One patient underwent cervicomedullary decompression surgery for chronic neck and back pain. These symptoms partially resolved initially but recurred one month post-operatively.

3.1.3 | Orthopedic Concerns

Thirty-eight patients (63%) had thoracolumbar kyphosis. In nearly all (37/38) cases, the kyphosis resolved without intervention as the patient aged and gained strength. In one patient, kyphosis was associated with symptoms of neurogenic claudication at 11 years of age; symptoms were not resolved at the time of data collection and corrective surgery was pending.

Fourteen patients (23%) had genu varum requiring surgical correction. Surgical correction resulted in com-

plete resolution of genu varum in 12 patients. In one patient, genu varum partially resolved following initial guided growth surgery, but subsequently recurred and was fully corrected after a second guided growth procedure. For one patient, the deformity was not adequately resolved with guided growth; therefore, an osteotomy was performed which fully corrected the genu varum.

3.1.4 | Respiratory Concerns

Fifteen patients (25%) had central sleep apnea, and 37 patients (62%) had obstructive sleep apnea. Among patients who underwent tonsillectomy/adenoidectomy, symptoms were fully resolved in 14 patients, partially resolved in seven patients, and not resolved in five patients. Outcome data following tonsillectomy/adenoidectomy were not documented for six patients.

Eleven patients (18%) were prescribed continuous positive airway pressure (CPAP) therapy. CPAP therapy resulted in complete resolution of symptoms in five patients, partial resolution in three patients, and no resolution in three patients.

3.1.5 | Auditory Concerns

Thirty-one patients (52%) experienced recurrent otitis media, 22 patients (37%) had middle ear effusions, and 23 patients (38%) had transient hearing loss. Twenty-nine patients (48%) underwent myringotomy tube placement. Indications for surgery included recurrent otitis media (22 patients), middle ear effusions (18 patients), hearing loss (14 patients), speech delay (seven patients), and nasal congestion (one patient); some patients had more than one indication. The indication for surgery was not specified for four patients.

Following myringotomy tube placement, symptoms were fully resolved in 13 patients, partially resolved in nine patients, and not resolved in two patients. Outcome data were not documented for five patients.

3.2 | Specialty Referrals

The most common specialty referrals included respirol-ogy (92%) for a sleep study; neurosurgery (75%) for foramen magnum stenosis; ENT (75%) for a tonsillectomy/adenoidectomy which is commonly used as a

treatment for obstructive sleep apnea; and orthopaedic surgery (67%) which is most commonly for an assessment of limb alignment. A complete list of specialties and referrals and the reasons underlying the referrals are summarized in supplementary Table S1.

4 | DISCUSSION

This retrospective study summarizes the clinical characteristics of children with achondroplasia who were evaluated within four years at SickKids to better understand the prevalence and management of achondroplasia-related comorbidities and concerns within the Canadian paediatric population. Fifty-two patients (87%) had a typical de novo mutation, keeping with a previously reported value of 80%. (10)

Concerning development, 50% of children with achondroplasia are reported to sit independently at nine months, (11) and the median age of walking independently in children with achondroplasia is 21.2 months. (12) We observed similar findings with an average age of sitting at 10 months and an average age of walking at 20 months. In our cohort, gross motor development milestones were available for only a subset of patients, with age at first sitting documented for approximately 20% and age at first walking for 58%. As a result, these findings should be interpreted cautiously and primarily as descriptive observations rather than representative estimates for the entire cohort. Gross motor delays are expected in children with achondroplasia due to biomechanical differences. Accordingly, achondroplasia-specific developmental charts should be used when assessing these patients. (12)

Excess weight is a significant risk factor for individuals with achondroplasia as it can exacerbate comorbidities such as obstructive sleep apnea, genu varum, spinal stenosis and increase mortality. (13) Twenty-four percent of our patients fell within the overweight or obese BMI category. Currently, no established standards exist to evaluate, prevent, or treat obesity in individuals with achondroplasia. (14) However, most patients in our study were counselled on healthy eating and exercise, whereas two were referred to nutrition/dietetics or obesity management. These findings suggest that

structured programs or recommendations may benefit children with achondroplasia who require weight management support. Overweight and obesity were more common in older patients in this cross-sectional study, suggesting that the study design may underestimate the true lifetime risk.

Neurological comorbidities were common in our study population, and 37% of the patients underwent cervicomedullary decompression surgery. This finding is higher than what has previously been reported (3-26%), (15,16) and may be attributed to the fact that Sick-Kids is a tertiary care center with a large neurosurgery department. The foramen magnum is small in children with achondroplasia, which can compress the brain stem and upper cervical spinal cord, potentially resulting in myelopathy, apnea, and sudden death. (17-19) Cervicomedullary decompression surgery may result in post-operatively complications such as cerebrospinal fluid leaks, although it has been found to provide significant clinical benefits in the context of achondroplasia. (20) Consistent with the literature, most of the patients in our study showed significant improvement post-surgery. No patients showed deterioration of symptoms or significant complications due to surgery.

Common orthopaedic concerns include thoracolumbar kyphosis (typically with an apex at the second lumbar vertebra) and genu varum. Thoracolumbar kyphosis usually resolves spontaneously as infants' muscle tone improves with age. (21) If the kyphosis does not resolve spontaneously, spine bracing may prevent progression, although its effectiveness is not proven. (21) A minority of paediatric patients may be considered for lumbar spinal surgery, particularly if there is a concomitant neurogenic claudication. (21) While 63% of patients in our study experienced kyphosis, only one patient (2%) required surgical correction. Joint hypermobility in the coronal plane of the knee is commonly observed in achondroplasia. This can exacerbate genu varum, initially caused by altered tibial shape. (22) It has been reported that approximately 10% of children have significant genu varum by age five, which increases to approximately 40% during adulthood. (23) This situation can lead to lateral knee pain and gait disruptions, includ-

ing a 'lateral thrust' when standing. Similarly, 23% of patients had genu varum that required surgical correction (guided growth, osteotomy, or limb lengthening). Surgical intervention was effective in reducing genu varum in our patient population. It is important to regularly assess for genu varum in patients with achondroplasia and consider surgical correction when patients have significant malalignment or asymptomatic malalignment before completing skeletal maturity. (24,25)

Approximately half of the patients (52%) in our study had episodes of recurrent otitis media, consistent with previous reports indicating that nearly 60% of patients experience at least one episode in the first year of life. (23) Conductive hearing loss has previously been reported in 38% of individuals with achondroplasia by adulthood. (23) We found that 38% of patients in our study showed evidence of transient conductive hearing loss. Auditory comorbidities may cause speech delays and articulation concerns. Of significance, myringotomy tube placement is effective in addressing these concerns. (26)

Sleep apnea is the most common respiratory problem in individuals with achondroplasia due to adenotonsillar hypertrophy or upper airway obstruction. (27) It has been reported that 25% of children with achondroplasia undergo tonsillectomy/adenoidectomy by eight years old and almost 40% by adulthood. (26) In the current study, 53% of patients underwent tonsillectomy/adenoidectomy. Reports of tonsillectomy/adenoidectomy effectiveness in treating obstructive sleep apnea have been mixed. One study reported that 60% of children treated with tonsillectomy/adenoidectomy required ongoing CPAP therapy to manage symptoms. (28) In contrast, another study reported that 64% of patients had complete resolution of obstructive sleep apnea symptoms after tonsillectomy/adenoidectomy and required no additional management. (29) We have observed that obstructive sleep apnea concerns were fully resolved following tonsillectomy/adenoidectomy in 38% of patients. Our findings highlight the need for ongoing monitoring and management of sleep apnea in this population, as some patients may require additional interventions to control obstruc-

tive sleep apnea fully.

4.1 | Strengths, Limitations, and Future Directions

While registries and natural history studies exist for achondroplasia, they are geographically limited to the United States, Europe, and India. (4,30,31) This study is the first to summarize the clinical findings of achondroplasia concerns and management within a Canadian paediatric population. This study also revealed the diverse and interrelated nature of medical concerns associated with achondroplasia which extend beyond stature and can cause significant morbidity. Pharmacological treatments for achondroplasia primarily focus on height and growth-related outcomes. (32) We recommend broadening the focus to also account for the concerns discussed above. Additionally, developing standardized methods to monitor and manage weight in children with achondroplasia may help to mitigate other serious comorbidities. Musculoskeletal pain, spinal pathology, and lower limb deformities may limit the feasibility of exercise-based weight management strategies in some patients with achondroplasia, underscoring the need for individualized, multidisciplinary approaches.

A limitation of this study is that there was limited availability of data on certain treatments and outcomes due to inconsistencies in reporting within patient charts, leading to potential gaps in the information collected. Our study offers valuable insight into the medical comorbidities and concerns of achondroplasia at SickKids, but its application may be limited as it only includes patients from a tertiary referral centre. Patients with fewer concerns may be less likely to attend referral centres, meaning estimates of medical event rates are likely higher from referral centre studies than from population-based studies. Individual variability in outcomes may be influenced by factors such as age, BMI, and baseline disease severity, which could not be systematically explored in this retrospective cohort.

As a retrospective chart review, outcome assessment relied on the accuracy and completeness of clinical documentation, which may introduce documentation or recall bias. Additionally, the duration of symptoms and

time to resolution could not be systematically evaluated due to inconsistent reporting. Distinguishing manifestations directly attributable to achondroplasia from those influenced by multifactorial contributors is challenging in retrospective studies and underscores the importance of cautious interpretation.

While most patients in our cohort harbored the typical *de novo* FGFR3 p.G380R variant, a small subset had inherited or atypical variants. Although genotype-phenotype correlations have been proposed in achondroplasia, the limited number of patients with non-classical variants in this cohort precluded meaningful subgroup analyses. Future multicenter registries with larger sample sizes may help elucidate whether specific genetic variants are associated with differential risks of particular clinical comorbidities and inform more personalized management strategies.

5 | CONCLUSION

This registry-based retrospective cohort study primarily aimed to characterize the range of medical and surgical concerns and their management in children with achondroplasia, rather than to evaluate the effectiveness of a registry itself. Our study highlights the need for various forms of medical and surgical management for children with achondroplasia by a multidisciplinary team. Health-care providers must be aware of these concerns, as early detection of comorbidities can be critical for optimal clinical outcomes. By improving our understanding of the clinical presentations involved in achondroplasia and what constitutes effective management, we can provide personalized care for this population of patients and improve their quality of life.

REFERENCES

1. Pauli RM. Achondroplasia: a comprehensive clinical review. *Orphanet J Rare Dis*. 2019;14(1):1. doi: 10.1186/s13023-018-0972-6
2. Bellus GA, Hefferon TW, Ortiz de Luna RI, Hecht JT, Horton WA, Machado M, et al. Achondroplasia is defined by recurrent G380R mutations of FGFR3. *Am J Hum Genet*. 1995;56(2):368–373.
3. Rousseau F, Bonaventure J, Legeai-Mallet L, Pelet A, Rozet JM, Maroteaux P, et al. Mutations in the gene encoding fibroblast growth factor receptor-3 in achondroplasia. *Nature*.

- 1994;371(6494):252–254. doi: 10.1038/371252a0
4. Hoover-Fong JE, Alade AY, Hashmi SS, Hecht JT, Legare JM, Little ME, et al. Achondroplasia natural history study (CLARITY): a multicenter retrospective cohort study of achondroplasia in the United States. *Genet Med*. 2021;23(8):1498–1505. doi: 10.1038/s41436-021-01165-2
5. Laederich MB, Horton WA. Achondroplasia: pathogenesis and implications for future treatment. *Curr Opin Pediatr*. 2010;22(4):516–523. doi: 10.1097/MOP.0b013e32833b7a69
6. Legare JM, Modaff P. Achondroplasia. In: GeneReviews® [Internet]. Seattle (WA): University of Washington; 1993–2025 [updated November 20, 2025; cited Dec 7, 2025]. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK1152/>
7. Fredwall SO, Åberg B, Berdal H, Savarirayan R, Solheim J. Hearing loss in Norwegian adults with achondroplasia. *Orphanet J Rare Dis*. 2021;16(1):468. doi: 10.1186/s13023-021-02095-7
8. Constantinides C, Landis SH, Jarrett J, Quinn J, Ireland PJ. Quality of life, physical functioning, and psychosocial function among patients with achondroplasia: a targeted literature review. *Disabil Rehabil*. 2022;44(21):6166–6178. doi: 10.1080/09638288.2021.196385
9. About SickKids. SickKids [Internet]. [Cited Dec 31, 2025]. Available from: <https://www.sickkids.ca/en/about/about-sickkids/>
10. Orioli IM, Castilla EE, Scarano G, Mastroiacovo P. Effect of paternal age in achondroplasia, thanatophoric dysplasia, and osteogenesis imperfecta. *Am J Med Genet*. 1995;59(2):209–217. doi:10.1002/ajmg.1320590218
11. Todorov AB, Scott Jr. CI, Warren AE, Leeper JD, Opitz JM. Developmental screening tests in achondroplastic children. *Am J Med Genet*. 1981;9(1):19–23. doi: 10.1002/ajmg.1320090105
12. Ireland PJ, Johnson S, Donaghey S, Johnston L, McGill J, Zankl A, et al. Developmental milestones in infants and young Australasian children with achondroplasia. *J Dev Behav Pediatr*. 2010;31(1):41–47. doi: 10.1097/DBP.0b013e3181c72052
13. Unger S, Bonafé L, Gouze E. Current care and investigational therapies in achondroplasia. *Curr Osteoporos Rep*. 2017;15(2):53–60. doi: 10.1007/s11914-017-0347-2
14. Saint-Laurent C, Garde-Etayo L, Gouze E. Obesity in achondroplasia patients: from evidence to medical monitoring. *Orphanet J Rare Dis*. 2019;14(1):253. doi: 10.1186/s13023-019-1247-6
15. Aryanpur J, Hurko O, Francomano C, Wang H, Carson B. Craniocervical decompression for cervicomedullary compression in pediatric patients with achondroplasia. *J Neurosurg*. 1990;73(3):375–382. doi: 10.3171/jns.1990.73.3.0375
16. Ryken TC, Menezes AH. Cervicomedullary compression in achondroplasia. *J Neurosurg*. 1994;81(1):43–48. doi: 10.3171/jns.1994.81.1.0043
17. Hecht JT, Nelson FW, Butler LJ, Horton WA, Scott Jr CI, Wassman ER, et al. Computerized tomography of the foramen magnum: achondroplastic values compared to normal standards. *Am J Med Genet*. 1985;20(2):355–360. doi: 10.1002/ajmg.1320200219
18. Nelson FW, Hecht JT, Horton WA, Butler LJ, Goldie WD, Miner M. Neurological basis of respiratory complications in achondroplasia. *Ann Neurol*. 1988;24(1):89–93. doi: 10.1002/ana.410240117
19. Pauli RM, Scott CI, Wassman ER, Gilbert EF, Leavitt LA, Ver Hoeve J, et al. Apnea and sudden unexpected death in infants with achondroplasia. *J Pediatr*. 1984;104(3):342–348. doi: 10.1016/S0022-3476(84)81092-6
20. Bagley CA, Pindrik JA, Bookland MJ, Camara-Quintana JQ, Carson BS. Cervicomedullary decompression for foramen magnum stenosis in achondroplasia. *J Neurosurg Pediatr*. 2006;104(3):166–172. doi: 10.3171/ped.2006.104.3.166
21. Pauli RM, Breed A, Horton VK, Glinski LP, Reiser CA. Prevention of Fixed, Angular Kyphosis in Achondroplasia. *J Pediatr Orthop*. 1997;17(6):726–733.
22. Kopits SE. Orthopedic aspects of achondroplasia in children. *Basic Life Sci*. 1988;48:189–197. doi: 10.1007/978-1-4684-8712-1.28
23. Hunter AG, Bankier A, Rogers JG, Sillence D, Scott CI. Medical complications of achondroplasia: a multicentre patient review. *J Med Genet*. 1998;35(9):705–712. doi: 10.1136/jmg.35.9.705
24. Kopits SE. Orthopedic complications of dwarfism. *Clin Orthop Relat Res*. 1976;(114):153–179.
25. Kopits SE. Genetics clinics of The Johns Hopkins Hospital. Surgical intervention in achondroplasia. Correction of bowleg deformity in achondroplasia. *Johns Hopkins Med J*. 1980;146(5):206–209.
26. Trotter TL, Hall JG, and the Committee on Genetics. Health supervision for children with achondroplasia. *Pediatrics*. 2005;116(3):771–783. doi: 10.1542/peds.2005-1440
27. Tasker RC, Dundas I, Laverty A, Fletcher M, Lane R, Stocks J. Distinct patterns of respiratory difficulty in young children with achondroplasia: a clinical, sleep, and lung function study. *Arch Dis Child*. 1998;79(2):99–108. doi: 10.1136/adc.79.2.99
28. Waters KA, Everett F, Sillence DO, Fagan ER, Sullivan CE. Treatment of obstructive sleep apnea in achondroplasia: evaluation of sleep, breathing, and somatosensory-evoked potentials. *Am J Med Genet*. 1995;59(4):460–466. doi: 10.1002/ajmg.1320590412
29. Sisk EA, Heatley DG, Borowski BJ, Levenson GE, Pauli RM. Obstructive sleep apnea in children with achondroplasia: surgical and anesthetic considerations. *Otolaryngol Head Neck Surg*. 1999;120(2):248–254. doi: 10.1016/S0194-5998(99)70414-6
30. Borkhuu B, Nagaraju DK, Chan G, Holmes LJ, Mackenzie WG. Factors related to progression of thoracolumbar kyphosis in children with achondroplasia: a retrospective cohort study of forty-eight children treated in a comprehensive orthopaedic center. *Spine*. 2009;34(16):1699–1705. doi: 10.1097/BRS.0b013e3181ac8f9d
31. Okenfuss E, Moghaddam B, Avins AL. Natural history of achondroplasia: a retrospective review of longitudinal clinical data. *Am J Med Genet A*. 2020;182(11):2540–2551. doi: 10.1002/ajmg.a.61825

32. Wrobel W, Pach E, Ben-Skowronek I. Advantages and disadvantages of different treatment methods in achondroplasia: a review. *Int J Mol Sci.* 2021;22(11):5573. doi: 10.3390/ijms22115573