CASE SERIES

Achondroplasia in the First Years of Life: Importance of Early Referral to Pneumology

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Abstract

Introduction: Achondroplasia is the most common bone dysplasia and is associated with extreme, disproportionate shortness of stature. Early care is essential to identifying risks and preventing serious sequelae. The American Academy of Pediatrics recommends conducting a polysomnography soon after the diagnosis of achondroplasia.

Methods: A retrospective study of children with achondroplasia referred to the multidisciplinary consultation in a central hospital in the first two years of life between 2015 and the end of the first semester of 2020.

Results: Ten children with achondroplasia were referred. The first evaluation in a pneumology consultation occurred between 3 and 22 months. Four children presented with snoring. The polysomnography was carried out between 5 and 26 months, revealing obstructive sleep apnea syndrome in seven children (including two with no snoring). The five with moderate/severe obstructive sleep apnea syndrome were started on non-invasive ventilation. All of them maintain non-invasive ventilation, most with good adherence. One of the children underwent an adenotonsillectomy at 2 years old and maintains ventilation. Magnetic resonance imaging of the brain showed the stenosis of the *foramen magnum* in all of the children. In the nine children who underwent polysomnography with no previous neurosurgical intervention, the rate of central apnea was normal.

Discussion: The prevalence and severity of obstructive sleep-disordered breathing in children with achondroplasia, especially in the first two years of life, justifies the early referral of these patients. Non-invasive ventilation was the most frequently used therapy and constitutes a valid and effective therapeutic option.

Keywords: Achondroplasia/complications; Child, Preschool; Infant; Polysomnography; Portugal; Treatment Outcome; Sleep Apnea, Obstructive/prevention & control; Sleep Apnea, Obstructive/therapy

Introduction

Achondroplasia is the most common form of bone dysplasia, which is associated with extremely short and disproportionate stature, presenting an incidence between 1/10,000 and 1/30,000 live births.[1,2] This is a genetic illness caused by a mutation in the recipient 3 of the fibroblast growth factor (*FGFR3*) gene of chromosome 4 (4p16.3), with autosomal dominant transmission, but in which 75%-80% of cases represent *de novo* mutations.[1,2]

Early referral of children with achondroplasia is essential to identifying the risks and intervening to prevent serious sequelae.[2] In this sense, children with achondroplasia have been followed since 2015, in the multidisciplinary bone dysplasia consultation at the Hospital Pediátrico – Centro Hospitalar e Universitário de Coimbra. This consultation aims to monitor children starting at the prenatal period, and includes genetics, endocrinology, pulmonology, neurosurgery, neuropediatric, physiatry, otorhinolaryngology (ENT), orthodontics, and orthopedics teams under pediatrics. As of 2017, this multidisciplinary team became part of the then established European Reference Network for Rare Bone Diseases (ERN-BOND).[3]

Respiratory problems during sleep are common in these children, particularly obstructive respiratory disorder, and pathological central apnea.

Obstructive respiratory sleep disorder, which includes obstructive sleep apnea syndrome (OSAS), is the most common respiratory complication, with a prevalence

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ranging from 22% to 93%.[4] The peculiarities of cranio-facial morphology, with hypoplasia of the central mass of the face, nasal bridge depression, ogival palate, and decreased mobility of the temporomandibular joint as well as hypotonia of the musculature of the upper airway lead to a greater risk of upper airway obstruction during sleep in these patients.[4,5]

Compression of the brainstem secondary to the stenosis of the *foramen magnum* may result in pathological central apnea and, although controversial, there seems to be a relationship between these events and the higher risk of sudden death in achondroplasiacs.[6]

The American Academy of Pediatrics recommends conducting a polysomnography study shortly after the diagnosis of achondroplasia.[2] The aim is to diagnose and treat the obstructive respiratory sleep disorder early on to avoid its consequences.

This study aims to characterize and provide therapeutic guidelines for respiratory sleep problems, targeting children with achondroplasia referred to the multidisciplinary consultation of bone dysplasia at the Hospital Pediátrico – Centro Hospitalar e Universitário de Coimbra during their first two years of life.

Methods

Retrospective, descriptive study, based on the clinical consultation of children with achondroplasia referred to the multidisciplinary consultation during the first two years of life between 2015 and the end of the first half of 2020.

The analyzed variables were sex, age at the start of the follow-up, current age, and age at which the polysomnography study (PSG) was performed as well as the presence of snoring and other respiratory symptoms at the first consultation growth (specific curves),[2] the result of cranioencephalic magnetic resonance imaging (CE-MRI), and the result of a polysomnography study, namely total sleep time (TST), efficiency, microarousal index (MAI), central apnea index (CAI), obstructive apnea/hypopnea index (OAHI), respiratory event related arousal (RERA) index, transcutaneous carbon dioxide ($TcCO_2$), peripheral oxygen saturation (SpO_2), and therapeutics.

The polysomnography studies were performed in the sleep and ventilation laboratory with the Alice 5 Respironics System®. The following signs were recorded: electroencephalogram (C3M2, C4M1, O1M2, O2M1, F3M2, F4M1), bilateral electrooculogram, submental electromyogram, right and left tibial electromyogram, electrocardiogram (D2), respiratory flow with nasal cannula and oronasal thermistor, thoracic and abdominal movements, peripheral oxygen saturation, transcutaneous carbon dioxide, respiratory noise, body position, and an audio-visual record.

The criteria defined by the American Association of Sleep Medicine were used in the polysomnography study analysis.[7] Sleep apnea syndrome was considered as mild for the obstructive apnea/hypopnea index between 1 and 4, moderate for the obstructive apnea/hypopnea index between 5 and 10, and severe for the obstructive apnea/hypopnea index > 10.[8] Upper airway resistance syndrome (UARS) was identified when the respiratory event related arousal index was > 1, without the criteria for sleep apnea syndrome. The pathological central apnea index was considered to be > 3.[9]

Results

Ten children with achondroplasia, including six males, were referred during the period of the study. The first assessment took place in a genetic consultation between the ages of 3 and 17 months (median age 6 months). All of the children had the typical phenotypic characteristics as hypoplasia of the midface, nasal bridge depression, ogival palate, and a receding position of the chin. At the end of the study, these children were aged between 7 and 64 months (five years).

The first pulmonology consultation took place between the ages of 3 and 22 months (median age of 5 months). Of the 10 children, four presented with snoring. No other significant respiratory symptoms were identified. None of the children presented poor weight progression, according to the growth curves adapted to achondroplasia. Table 1 shows the demographic and clinical characteristics, polysomnography study, CE-MRI results, and the therapeutic guidelines of these children.

The polysomnography study was performed between the ages of 5 and 26 months (median age of 9 months), with an efficiency that varied between 65% and 91% (median 78%). Table 2 shows the respiratory parameters analyzed in the polysomnography study.

Regarding diagnoses, seven children were diagnosed with obstructive sleep apnea syndrome, four severe, one moderate, and two mild. The obstructive apnea/hypopnea index ranged from 2.8 to 40.5, the minimum SpO_2 from 61% to 90%, and the maximum $TcCO_2$ from 38-65 mmHg. Two of the children with severe sleep apnea syndrome did not present snoring. The five children with moderate to severe sleep apnea syndrome started non-invasive ventilation (NIV) during hospitalization, with good adaptation, between the ages of 7 and 29 months.

The most applied ventilation mode was continuous positive airway pressure (CPAP), with pressures ranging from 4 to 7 cm H_2O. All of them maintain non-invasive ventilation, most with good adherence and a median duration of 18 months (3-25 months).

In one of the children, the polysomnography study showed upper airway resistance syndrome, with a respiratory event related arousal index of 5.7, a minimum SpO_2 of 87%, and a maximum $TcCO_2$ of 40 mmHg.

Children with mild sleep apnea syndrome and upper airway resistance syndrome were started on medical therapy with nasal corticosteroids and montelukast.

Children who were not under non-invasive ventilation (normal polysomnography study, with mild sleep apnea syndrome or upper airway resistance syndrome) were monitored with a semi-annual or annual polysomnography study. Only one of the children with a previously normal polysomnography study demonstrated a change in the result, and mild sleep apnea syndrome was identified.

All of the children were referred to otorhinolaryngology. One of the children underwent adenotonsillectomy at 31 months, presenting with severe sleep apnea syndrome on the polysomnography study, under non-invasive

Table 1. Clinical and demographic characteristics, polysomnography study result, cranioencephalic magnetic resonance imaging result and guidelines

	Gender	Weight/height ratio	Snoring	Age PSG	PSG	Therapeutic guidelines	CE-MRI NS	ENT surgery	Current age
Case 1	M	P15	No	5 m	Mild OSAS	Monitoring	Stenosis foramen magnum with spinal cord compression Surgery 7 m	-	6 m
Case 2	M	P15	Yes	5 m	Severe OSAS	NIV - CPAP	Stenosis foramen magnum Monitoring	-	10 m
Case 3	F	P50	No	9 m	Severe OSAS	NIV - BiPAP	Stenosis foramen magnum Monitoring	-	40 m
Case 4	F	P15-50	No	9 m	Normal	Monitoring	Stenosis foramen magnum Monitoring	-	36 m
Case 5	M	P50-75	Yes	7 m	Severe OSAS	NIV - CPAP	Stenosis foramen magnum Monitoring	-	10 m
Case 6	M	P3	No	7 m	Mild OSAS	Monitoring	Stenosis foramen magnum with spinal cord compression and hydrocephaly Surgery 9 m	-	11 m
Case 7	M	P50	No	17 m	Severe OSAS	NIV - CPAP	Stenosis foramen magnum with spinal cord compression Surgery 10 m	-	39 m
Case 8	M	P15	Yes	22 m	UARS	Monitoring	Stenosis foramen magnum Monitoring	-	64 m
Case 9	F	P3-15	No	26 m	Normal	Monitoring	Stenosis foramen magnum Monitoring	-	49 m
Case 10	F	P3-15	Yes	25 m	Moderate OSAS	NIV - CPAP	Stenosis foramen magnum Monitoring	T&A 31 m	54 m

BiPAP - bilevel positive airway pressure; CE-MRI - cranioencephalic magnetic resonance imaging; CPAP - continuous positive airway pressure; F - female; M - male; m - months; NIV - non-invasive ventilation; NS - neurosurgery; OSAS - obstructive sleep apnea syndrome; PSG - polysomnography study; UARS - upper airway resistance syndrome; T&A - adenoidectomy + tonsillectomy.

Table 2. Parameters analyzed in the polysomnography study

	TTS (min)	Efficiency (%)	MAI	OAHI	CAI	RERAI	Average SpO_2 (%)	Minimum SpO_2 (%)	$TcCO_2$ maximum (mmHg)	PSG result
Case 1	519	80	14.4	4.3	0.5	1.5	97	90	---	Mild OSAS
Case 2	446	76	9	12.4	1.6	0.3	98	88	52	Severe OSAS
Case 3	532	87	12.5	14	1.1	0	95	76	42	Severe OSAS
Case 4	542	73	6	0	1.8	0.1	97	81	41	Normal
Case 5	466	71	22.8	40.5	0	25.6	96	61	65	Severe OSAS
Case 6	475	80	15.7	2.8	2.7	1.6	97	87	47	Mild OSAS
Case 7	526	91	17.4	14.8	0	4.1	98	93	55	Severe OSAS
Case 8	440	72	17.9	0.1	0.9	5.7	99	87	40	UARS
Case 9	383	65	11.9	0.6	0	0.1	98	95	55	Normal
Case 10	514	87.6	11.9	5.4	0.8	0.6	95	74	38	Moderate OSAS

CAI - central apnea index; MAI - microarousal index; OAHI - obstructive apnea/hypopnea index; OSAS - obstructive sleep apnea syndrome; RERAI - respiratory event related arousal index; SpO_2 - peripheral oxygen saturation; $TcCO_2$ - transcutaneous carbon dioxide; TST - total sleep time; UARS - upper airway resistance syndrome.

ventilation, but with poor adhesion due to persistent nasal obstruction and repeated high respiratory infections. After adenotonsillectomy, the patient maintained the need for non-invasive ventilation.

The CE-MRI showed stenosis of the *foramen magnum* in all children followed by consultation. Three of these children underwent surgery for decompression of the *foramen magnum*, one of them after undergoing a polysomnography study. In all other (nine), the central apnea index was normal, ranging from 0-2.7.

Only three children were already observed in a neurodevelopment consultation, and none of them were found to have delayed psychomotor development.

Discussion

Achondroplasia is the most common form of bone dysplasia, with internationally defined guidelines for monitoring starting from the prenatal period. These guidelines aim to anticipate and guide the issues related with this pathology, to minimize the risk of complications and sequelae. At our hospital, as of 2015, children with achondroplasia are followed up in a multidisciplinary bone dysplasia consultation. This study aimed to characterize the respiratory sleep problems and therapeutic guidelines of children with achondroplasia during the first two years of life, followed through this consultation.

In children with achondroplasia, polysomnography study is indicated as soon as the diagnosis is established, even in the absence of respiratory sleep disorder symptoms.[2] In our sample, the polysomnography study was performed between 5 and 26 months of age, a consequence not only of the age at which the children were referred, but also of the difficulties and limitations related to performing this auxiliary test, particularly in a pediatric setting.

The diagnosis of sleep apnea syndrome was very frequent, and it was present in 7 of the 10 children in the study. Apart from snoring, no other associated symptoms were identified, namely other respiratory symptoms or poor weight progression. It should also be noted that, in two of the children with severe sleep apnea syndrome, there was no snoring or any other symptom suggestive of an obstructive respiratory sleep disorder. These data are in accordance with what has been described in the literature, where a higher probability of sleep apnea syndrome at the age of 3 or less is demonstrated, notably it the possible absence of symptoms, which may be silent or even underestimated by parents.[4,6]

In children with achondroplasia, the etiology of obstructive respiratory disorder is multifactorial, and is related to the consequences of bone growth anomaly in the cranial base and face, and to the risk factors for this disorder in pediatric age groups. The characteristics of facial morphology, such as hypoplasia of the central mass, nasal bridge depression, ogival palate and the receding position of the chin, lead to an increase in the mandibular angle and length of the lower third of the face, which contribute to the narrowing of the upper airways in these patients.[4] In addition to these structural factors, there may also be hypotonia of the musculature as a consequence of stenosis of the jugular foramen and the hypoglossal nerve.[6] The role of adenotonsillar hypertrophy, as the most common etiological factor for obstructive respiratory disorder in pediatric sleep, becomes determinant from the age of 2.[5]

Considering this multifactorial etiology, the treatment used in children with moderate to severe sleep apnea syndrome was non-invasive ventilation. At our hospital, the adaptation to non-invasive ventilation is always done under hospitalization, a factor that, in the authors experience, has proven to be determinant in treatment adherence at these ages. At the time of completion of this study, all of the children (aged between 10 months and 4 years) maintained non-invasive ventilation. Even so, the need for non-invasive ventilation is expected to be transient, given the natural history of obstructive respiratory sleep disorder in these children. With growth, the probability of obstructive apnea decreases, as demonstrated in a study of 43 children with achondroplasia, where all of the children with apnea/hypopnea index ≥ 10/h were younger than 7 years of age.[5]

In our sample, only one child underwent adenotonsillectomy, and it was necessary to maintain non-invasive ventilation after the otorhinolaryngology surgery. The latter observation agrees with the multifactorial etiology of obstructive respiratory disorder in these children, and the fact that adenotonsillar hypertrophy is mostly preponderant starting at 2 years of age. Therefore, as described in other studies, otorhinolaryngology surgery is not the main therapeutic option under the age of 2.[10]

Abnormal chest wall growth and progressive kyphoscoliosis can contribute to the development of restrictive lung disease, occurring in less than 5% of children younger than 3 years old.[6] Respiratory follow-up is needed to diagnose this consequence of achondroplasia early.

Although all of these children had a stenosis of the *foramen magnum*, three with medullary involvement,

the rates of central apnea in their polysomnography study were normal. In addition, no cases of sudden death were verified. Classically, central apnea in children with achondroplasia is described as the result of dysfunction of the respiratory centers by compression of the brainstem or upper spinal cord, combined with the premature fusion of the occipital segments.[6,11] The possible occurrence of lethal central apnea, as a cause of death among infants with achondroplasia, is estimated at 7.5% during the first year of life and 2.5% between 1 and 4 years of age.[6,11] The absence of central apnea in our study is in agreement with the findings of a study including 46 children with achondroplasia, in which the median central apnea index was zero.[10] The relationship between apnea and stenosis of the *foramen magnum* remains controversial and, therefore, polysomnography study findings should not be interpreted in isolation when considering neurosurgical intervention.[6,11,12]

Achondroplasia is not in itself associated with intellectual development disorder.[2] However, the issues related with this pathology, particularly breathing disorders during sleep, can contribute to delayed psychomotor development or learning disabilities.[2] Obstructed breathing during sleep is associated with neurocognitive, behavioral, cardiovascular, and metabolic consequences.[13,14] In the pediatric age, neurocognitive and behavioral consequences are of particular relevance, and psychomotor development delay, learning disabilities, attention deficits and hyperactivity, mood disorders and anxiety, and defiant behaviors have been described, among others.[14] Early diagnosis and treatment of respiratory sleep disorder is, therefore, key to avoiding these consequences. There were no changes in psychomotor development in our sample, which may be due to early follow-up or the evaluation time being only up to 5 years.

The median age of referral to the bone dysplasia multidisciplinary consultation at our hospital was 6 months. Currently, the international guidelines for the follow-up of these children recommend referral from the prenatal period, to plan all the complementary research during the first months of life, namely polysomnography study and central nervous system imaging. In this manner, the intent is to anticipate the age of referral, through better articulation between obstetric and pediatric teams.

Our study has some limitations, particularly the fact that it is a retrospective study with a small patient sample, inherent to the rarity of this pathology. We also highlight the difficulty in performing polysomnography study as early as the first months of life, given the age at which patients are referred, and the existing limitations for performing this type of test.

The prevalence and severity of obstructive respiratory sleep disorder in children with achondroplasia, particularly during the first two years of life, justify early referral for these patients. At this age, the use of non-invasive ventilation is a valid and effective therapeutic option. We highlight the importance of multidisciplinary assessment and follow-up for these children at experienced centers, with referral carried out, if possible, during the prenatal period or immediately after birth.

In Portugal, bone dysplasia has not yet been selected as an area for the formal establishment of referral centers.[15]

WHAT THIS STUDY ADDS

- This study presents the first results of respiratory follow-up of children with achondroplasia after the implementation of a multidisciplinary consultation of bone dysplasia in a hospital in Portugal.
- This is the first study that includes achondroplasics exclusively in their early years of life.
- Moderate to severe obstructive sleep apnea syndrome is more common in these early years, but it does not necessarily lead to snoring evidence.
- Noninvasive ventilation is the treatment of choice for obstructive apnea syndrome in children with achondroplasia of this age group.
- The creation of this multidisciplinary consultation and these preliminary results may contribute to the future selection of bone dysplasia as areas for the formal establishment of referral centers in Portugal.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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Consent for publication

Consent for publication was obtained.

Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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Acondroplasia nos Primeiros Anos de Vida: Importância da Referenciação Precoce à Pneumologia

Resumo

Introdução: A acondroplasia é a displasia óssea mais comum e está associada a uma baixa estatura extrema e desproporcional. Cuidados médicos precoces são essenciais para identificar riscos e prevenir sequelas graves. A American Academy of Pediatrics recomenda a realização de uma polissonografia logo após o diagnóstico de acondroplasia.

Métodos: Estudo retrospetivo de crianças com acondroplasia referenciadas para consulta multidisciplinar num hospital central nos primeiros dois anos de vida, entre 2015 e o final do primeiro semestre de 2020.

Resultados: Foram referenciadas dez crianças com acondroplasia. A primeira avaliação em consulta de pneumologia ocorreu entre 3-22 meses. Quatro crianças ressonavam. A polissonografia foi realizada entre os 5-26 meses, revelando síndrome da apneia obstrutiva do sono em sete crianças (incluindo duas que não ressonavam). As cinco crianças com síndrome da apneia obstrutiva do sono moderada / grave iniciaram ventilação não invasiva. Todas mantêm ventilação não invasiva, a maioria com boa adesão. Uma das crianças foi submetida a adenoamigdalectomia aos 2 anos e mantém ventilação. A ressonância magnética crânio-encefálica revelou estenose do forame magno em todas as crianças. Nas nove crianças que fizeram polissonografia sem intervenção neurocirúrgica prévia, a taxa de apneia central foi normal.

Discussão: A prevalência e a gravidade dos distúrbios respiratórios obstrutivos do sono em crianças com acondroplasia, principalmente nos dois primeiros anos de vida, justificam a sua referenciação precoce. A ventilação não invasiva foi o tratamento mais utilizado e constitui uma opção terapêutica válida e eficaz.

Palavras-Chave: Acondroplasia/complicações; Apneia Obstrutiva do Sono/prevenção & controle; Apneia Obstrutiva do Sono/tratamento; Criança; Lactente; Polissonografia; Portugal; Resultado do Tratamento