Spine Deformity
www.spine-deformity.org

Spine Deformity 4 (2016) 145−148

Prevalence of Scoliosis and Thoracolumbar Kyphosis in Patients With Achondroplasia

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Received 8 January 2015; revised 6 August 2015; accepted 8 August 2015

Abstract

Study Design: Retrospective chart review, case series.
Objectives: To determine the prevalence of scoliosis and kyphosis in patients with achondroplasia.
Summary of Background Data: There is little published research on the prevalence of scoliosis and thoracolumbar kyphosis in patients with achondroplasia.
Methods: The authors retrospectively reviewed charts of 459 patients with achondroplasia who were seen by the senior author, an orthopedic surgeon, from 1999 through 2013, at a tertiary referral center. After excluding patients who presented after spinal surgery and those who were referred for specific non-spinal issues, 326 patients were included (71%). Cobb angles were measured on lateral and posteroanterior radiographs. Scoliosis was defined as curvature on posteroanterior radiographs greater than 10°; thoracolumbar kyphosis was defined as any kyphotic curvature with an apex between T11 and L2. These data were then stratified by sex, age group (0−2, 3−12, 13−19, 20−40, and >40 years), and severity: within normal limits (≤10°), mild (>10°−25°), moderate (26°−50°), and severe (>50°).
Results: The study population consisted of 176 males and 150 females with a mean age of 18 years. Scoliosis was observed in 60%. Thoracolumbar kyphosis was observed in 79%, with 52% exhibiting moderate to severe curvature.
Conclusions: In these patients, the rates of scoliosis and kyphosis were 60% and 79%, respectively, which are much higher than the rates reported in the literature for the general population of children.
Level of Evidence: Level 3 or 4.

Keywords: Achondroplasia; Kyphosis; Prevalence; Scoliosis; Skeletal dysplasia

Introduction

Though relatively rare, achondroplasia is the most common skeletal dysplasia, affecting 1 in 30,000 live births annually [1]. This disorder, caused by a mutation in the fibroblast growth factor receptor 3 gene, presents in infancy as a distinct form of rhizomelic short stature.

Patients with achondroplasia present with mild to severe abnormalities of the spine, including thoracolumbar kyphosis, scoliosis, and spinal stenosis. Progression of spinal involvement follows a characteristic timeline in most patients, with many newborns having radiographic findings consistent with thoracolumbar kyphosis [2,3]. As children progress from sitting to standing and beginning to ambulate, kyphosis decreases significantly. However, by age 50, rates of kyphosis rise again [4].

Fifteen percent of adults with achondroplasia have kyphosis with neurologic sequelae [1], and much research has focused on determining factors that result in progression of thoracolumbar kyphosis. Studies have shown a connection between spinal curvature deformities and other clinical manifestations. Hypotonia, hydrocephalus, and delayed motor development have been shown to worsen kyphosis in this population [1,4,5]. Yet, despite a clear association between kyphosis and the development of

Author disclosures: BIK (none), MTY (none), HB (none), MCA (none).

IRB approval for this study was obtained from the institutional review board in the appropriate manner, before the collection or analysis of any patient data, and the study was conducted fully abiding by the rules and regulations set out by the review committee.

This study received approval from The Johns Hopkins University institutional review board. Reference number, CR00001242.

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2212-134X/$ - see front matter © 2016 Scoliosis Research Society.
http://dx.doi.org/10.1016/j.jspd.2015.08.003

neurologic symptoms, as well as a clear role for early intervention in certain cases, there are scant data on the prevalence of thoracolumbar kyphosis and scoliosis in patients with achondroplasia.

This study aims to quantify the prevalence of scoliosis and kyphosis in patients with achondroplasia by analyzing a large database of records of patients seen over 14 years at a tertiary care center.

Materials and Methods

Over 14 years, 541 patients with achondroplasia were seen by the senior author. Of these, 82 patients were excluded because they were last seen more than 5 years before the implementation of an updated electronic medical record system, and thus their records were unavailable. A retrospective chart review was performed on the remaining 459 patients.

Of these 459, a total of 95 patients were excluded because they presented to our clinic after spinal surgery. Additionally, 38 patients were referred for specific non-spinal complaints and thus did not undergo scoliosis studies. This left 326 patients for inclusion in our study.

All Cobb angles were measured on lateral and posteroanterior radiographs by one individual to prevent interobserver bias. Cobb angles were analyzed using standard electronic image visualizing software (UltraVisual, Merge Healthcare, Chicago, IL). Scoliosis was defined as any curvature greater than 10°. Kyphosis, though usually normal up to 40° in the thoracic region, should be minimal, if at all present, at the thoracolumbar junction [6]. Therefore, for this study, a conservative cutoff of greater than 10° kyphotic curvature at the thoracolumbar junction, with apex between T11 and L2, was used as diagnostic for thoracolumbar kyphosis. Data were stratified by sex, age group (0–2, 3–12, 13–19, 20–40, and >40 years), and severity of curvature (within normal limits [≤10°], mild [>10°–25°], moderate [26°–50°], and severe [>50°]).

Scoliosis and kyphosis were analyzed separately in each patient and stratified by age, sex, and severity of curvature. Data were analyzed with Microsoft Excel software (Microsoft Corp., Redmond, WA) using simple univariate analysis to determine prevalence and correlation of kyphosis to scoliosis.

Results

After exclusion criteria were applied, 326 patients (176 males and 150 females) were eligible for inclusion. The mean age, at time of radiographs, was 18.0 years.

Of these patients, 60% had some degree of scoliosis (Table 1), with 6.1% having moderate to severe curvature (>25°). No differences were noted by sex (60% males, 60% females) or age group. Thoracolumbar kyphosis, using the diagnostic cutoff of greater than 10° kyphotic curvature at the thoracolumbar junction with apex between T11 and L2, was observed in 79% of patients (Table 2), with 52% having moderate to severe curvature (>25°). The majority of our patient population had kyphotic curvature between T8 and L3. As with the scoliosis measurements, there were minimal differences by age group, with a slightly higher rate of kyphosis in males than females (87% vs 70%).

Table 1
Prevalence of scoliosis in a population of patients with achondroplasia, by age group.

Age group and scoliosis severity	Males (n = 176)		Females (n = 150)	
	No.	%	No.	%
0–2 years				
WNL[a]	14	41.18	13	50.00
Mild[b]	19	55.88	13	50.00
Moderate[c]	1	2.86	0	0.00
Severe[d]	0	0.00	0	0.00
Total	34		26	
3–12 years				
WNL	35	46.05	16	39.02
Mild	40	52.63	21	51.22
Moderate	1	1.32	3	7.32
Severe	0	0.00	1	2.44
Total	76		41	
13–19 years				
WNL	10	37.04	11	45.83
Mild	17	62.96	11	45.83
Moderate	0	0.00	1	4.17
Severe	0	0.00	1	4.17
Total	27		24	
20–40 years				
WNL	8	36.36	10	47.62
Mild	11	50.00	10	47.62
Moderate	2	9.09	1	4.76
Severe	1	4.55	0	0.00
Total	22		21	
>40 years				
WNL	3	17.65	10	26.32
Mild	11	64.71	23	60.53
Moderate	3	17.65	4	10.53
Severe	0	0.00	1	2.63
Total	17		38	

WNL, within normal limits.
[a] WNL, ≤10° curvature.
[b] Mild, >10°–25° curvature.
[c] Moderate, 26°–50° curvature.
[d] Severe, >50° curvature.

A comparison of the degree of kyphotic curvature with the degree of scoliosis showed no correlation between the extent of scoliosis and the extent of kyphosis within individual patients (correlation coefficient = 0.20) (Fig. 1).

Discussion

Analysis of posteroanterior and lateral radiographs of these patients with achondroplasia showed a prevalence of 60% for scoliosis and 79% for thoracolumbar kyphosis. Not surprisingly, these rates are much higher than those reported for the general population [7,8]. Adolescent