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**CLINICAL CASE** 

# Lumbar hypoplasia associated to thoracolumbar kyphosis in infants. Anatomic variant? Clinical cases

Hipoplasia vertebral asociada a cifosis toracolumbar en lactantes ;variante anatómica? Casos clínicos

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#### **Abstract**

**Introduction**: Vertebral hypoplasia is an anomaly of the vertebral bodies, in which they present a wedge shape, usually at the level of the thoracolumbar junction. Although it is associated with certain storage diseases and bone dysplasias, it is also possible to find isolated vertebral hypoplasia it in healthy infants or associated with thoracolumbar kyphosis. The objective of this report is to show the evolution of vertebral hypoplasia associated to kyphosis in two apparently health children. **Case Report**: Two cases of infants diagnosed with clinically visible lumbar kyphosis when they were sitting. Spine X-rays of both showed lumbar vertebral hypoplasia at L2 level as the only finding. After ruling out other conditions associated with vertebral hypoplasia, conservative management was indicated; in the first case a clinical-radiological follow-up and in the second one, a corset given the magnitude of kyphosis. The evolution was favorable, with complete radiological clinical resolution at the age of 15 months in the first case and clinical regression in the second, in which, at 3 years and 4 months of age, an image of mild vertebral hypoplasia persisted. **Conclusions**: Isolated vertebral hypoplasia or associated to kyphosis may be considered a minor anomaly or anatomic variant of infant spine development, however, it requires follow-up until its normalization.

# **Keywords:**

Kyphosis; spine deformities; vertebral hypoplasia; lumbar vertebrae

## Introduction

Congenital anomalies of the spine are rare in pediatrics, and can be divided according to their origin, into malformations of the medullar-radicular axis or spinal dysraphia that can be open or closed and malformations of the vertebrae, whether isolated or associated with other bone abnormalities of the spine, skin, neurological and/or visceral syndromes<sup>1</sup>.

The anomalies of the vertebrae are further classified into segmentation, formation or mixed defects. The alteration in the vertebral segmentation corresponds to the presence of a bone bar or block of two or more vertebrae, at anterior, posterior or lateral level. When the anterior vertebral formation fails bilaterally, aplasia or vertebral hypoplasia is generated, and if the defect is unilateral, a hemivertebra is developed<sup>1</sup>.

To better understand the structural anomalies of the spine, it should be considered that the Endochondral ossification of the vertebral body begins in fetal development at the thoracolumbar level and from there it progresses towards cephalic and caudal, being completed at the age of 18 to 20 years2. Due to the different ossification rhythms of different portions of the vertebrae, it is common to find anatomical variants of the vertebral bodies during childhood; Gaca et al. described that in 95% of children, in the vertebrae at thoracolumbar junction level, the relationship between anterior and posterior wall height (A/P ratio), is higher than 89.3% without showing differences according to age<sup>3</sup>; however, when the A/P difference is less than 89% and especially if there is an anterior wedge shape, it is suggestive of an injury of the vertebral body, or a morphological alteration or vertebral hypoplasia<sup>4</sup>.

In parallel with the development of the vertebrae, as the gross motor progresses, the curvatures of the spine in the sagittal plane are physiologically modified. At birth, a primary kyphosis predominates throughout the entire spine, being the point of greatest stress at the thoracolumbar junction level. As the infant begins with the cephalic support and prone head lift, secondary curvatures develop, such as cervical lordosis and then, in relation to standing, lumbar lordosis<sup>5</sup>, with kyphosis not normally found at this level.

Thoracolumbar vertebral hypoplasia occurs more frequently in children affected by skeletal dysplasia or storage diseases<sup>6</sup>. In healthy children, isolated vertebral hypoplasia is usually an incidental finding on chest or spine x-rays<sup>7,8</sup>, although it may be associated with alterations in the longitudinal growth of a segment of the spine, determining the development of kyphosis or kyphoscoliosis<sup>9</sup>.

This finding, although it could correspond to a minor anomaly or anatomical variant of good prognosis, generates great anguish in parents and health team. The objective of this report is to show the evolution of vertebral hypoplasia associated with kyphosis in two apparently healthy infants.

#### Clinical cases

#### Case 1

A female infant with a history of food allergy and gastroesophageal reflux, with no other relevant morbid or perinatal history. At nine months of age, her parents consulted for a lumbar prominence, which they had noticed since 4-5 months of age. Her psychomotor development had always been adequate; the parents reported not having sat her before six months of age, spending much time in prone position. Physical examination revealed no dysmorphia or disproportionate body segments and the anthropometric assessment was adequate for the age.

Anteroposterior and lateral x-ray of the spine taken at nine months of age showed thoracolumbar kyphosis of 18° (according to Cobb method<sup>10</sup>) associated with vertebral hypoplasia of the anterosuperior side of the lumbar vertebra L2, with an A/P ratio of 0.86 (Figure 1). Clinically and radiologically there were elements of suspicion of secondary causes. Evaluated by pediatric traumatology, clinical and radiological observation and control were recommended.

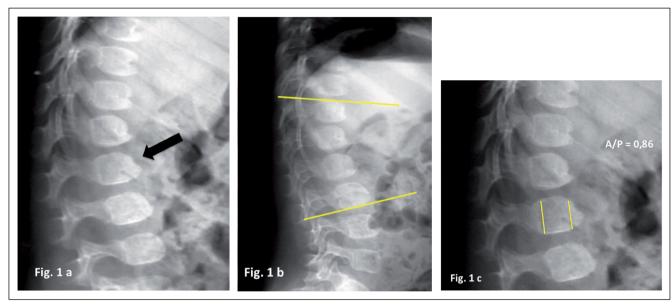
At one year of age, the patient had an adequate psychomotor development and a good pondoestatural growth. Physical examination did not show kyphosis. The follow-up chest X-ray, at 15 months of age, showed the normalization of the vertebral wedge and kyphosis. In this test, the A/P ratio was 0.88 (Figure 2).

## Case 2

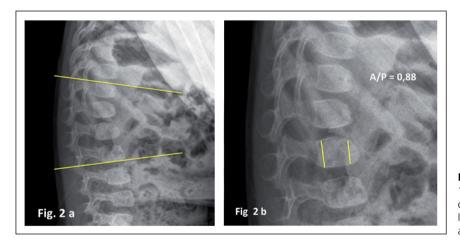
Male infant, with no significant morbid, family or perinatal history. In a well-baby visit at 5 months of age, the presence of a lumbar prominence was investigated. Asking parents in a directed way, they reported having noticed a small deformity at that level since 2-3 months of age, being this more pronounced when sitting.

The infant had a normal psychomotor development, in the remote anamnesis, it was not possible to define the time that spent in prone. Physical examination revealed no dysmorphia or disproportionate body segments and the anthropometric evaluation was appropriate for the age according to WHO reference<sup>11</sup>, always maintaining an adequate growth rate.

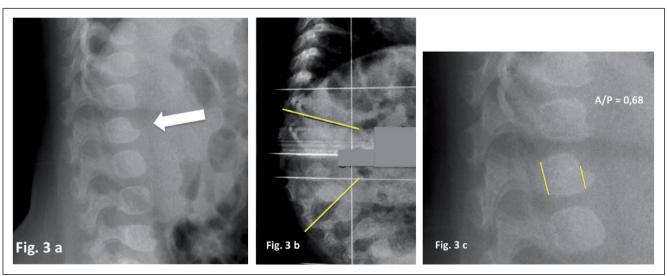
The lateral X-ray of the spine with the child seated showed thoracolumbar kyphosis of 57° (according to the Cobb method<sup>10</sup>) associated with vertebral hypoplasia of the anterosuperior portion of the L2 vertebra. The A/P ratio was 0.68 (Figure 3). Clinically and radiologically there were no suspicions of mucopolysac-



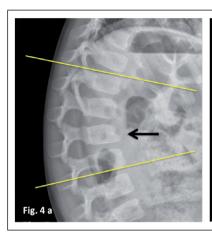
**Figure 1.** Case 1: Lateral radiograph of the spine at age 9 month, shows hypoplasia of the anterosuperior margin of L2 (**a**, arrow). Standing lateral radiograph (**b**) demonstrates 18° kyphosis (Cobb). L2 anterior and posterior height ratio of 0.86.

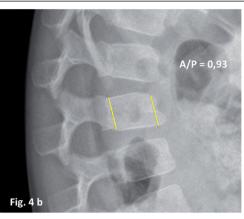


**Figure 2.** Case 1: Follow up radiograph at age 15 month, lateral sitting proyection (accentuation of dorsal kyphosis) demonstrate 18° kyphosis (**a**). Interval decrease of the L2 hypoplasia. L2 anterior and posterior height ratio of 0.88 (**b**).



**Figure 3.** Case 2: Lateral radiograph at age 5 month. Noticeable anterosuperior L2 hypoplasia (arrow in **a**) Lateral sitting position radiograph shows pronounced dorso lumbar 57° kyphosis (**b**) L2 anterior and posterior height ratio of 0.68 (**c**).





**Figure 4**. Case 2: Lateral sitting position radiograph at age 3 years and 4 month, demostrate slight 25° kyphosis (**a**), persistent anterosuperior hypoplasia of L2 (arrow). L2 anterior and posterior height ratio of 0.93 (**b**)

charidosis, achondroplasia, hypothyroidism, or other secondary causes such as trauma, hypotonia, or infections. Due to the important degree of angulation, the use of corset was indicated, which he used for 23 hours a day between eight and 26 months of age.

The infant began to walk at 14 months, with adequate acquisition of the milestones of psychomotor development and growth rate according to expectations. The last evaluation, conducted at three years and four months of age, showed that kyphosis had evolved favorably. On that occasion, the anthropometric evaluation was also normal. The X-ray showed a kyphosis of 25° in a seated position, however, a slight hypoplastic aspect of the anterosuperior segment of the lumbar vertebra L2 continued, which was not observed in the A/P ratio of 0.93 (Figure 4). Due to the persistence of mild kyphosis, it was decided to continue with the use of the corset.

#### Discussion

Childhood kyphosis can be congenital or acquired, such as those caused by trauma, sepsis or hypotonia<sup>1</sup>. Kyphosis associated with vertebral hypoplasia has been described in children affected by skeletal dysplasia such as achondroplasia, hypochondroplasia, and rhizomelic chondrodysplasia punctata in storage diseases, for instance, mucopolysaccharidosis and in endocrinological diseases such as congenital hypothyroidism<sup>6,12</sup>. In our patients, there were no elements of clinical suspicion of disease associated at the time of diagnosis, which added to a favorable evolution, ruled out a causal etiology. Considering that isolated vertebral hypoplasia or associated with kyphosis has been identified in healthy children<sup>7,13,14</sup>, it has been discussed in these cases as an anatomical variant of the spinal development<sup>8</sup>.

The degree of kyphosis is measured through the Cobb method<sup>10</sup> which was originally described for its application in idiopathic scoliosis in adolescents. In infants and preschool children with greater musculoskeletal laxity, this measurement is less useful and may overestimate kyphosis curves at these ages, especially if measured with the patient seated. Another way to assess the morphology of the vertebrae is to measure the height in the sagittal plane of the vertebral body in its anterior and posterior walls, determining a ratio between both measures: the expected A/P ratio is about 89%<sup>3</sup>. The limitation of this measurement is that there may be inter and intra-observer variability, which makes it difficult to investigate vertebral anomalies<sup>15</sup>.

The first description of kyphosis associated with vertebral hypoplasia was made by L. Swischuk in 1970, attributing this alteration to hypotonia, exaggeration of the normal kyphosis of the thoracolumbar area, anterior herniation of the nucleus pulposus and anterior subluxation of the vertebral body<sup>16</sup>. Other authors have associated it with mechanical factors that favor a nonphysiological curvature of the spine, such as excessive sitting in infants who have not developed paravertebral musculature and lack of prone posture<sup>13</sup>. However, it is not possible to determine if vertebral hypoplasia is secondary to vertebral compression that occurs in the thoracolumbar junction, which is the point of greatest inflection in a kyphosis; or if it is a primary anomaly of the vertebra and this determines the development of kyphosis. In this regard, any factor that interferes with the longitudinal growth of the anterior portion of the vertebral body should be considered to determine local angulation2.

The diagnosis of isolated vertebral hypoplasia or associated with kyphosis is based on the radiological appearance of the vertebrae, the lack of anomalies in posterior elements, the absence or minimal anterior

	Campos et al. (2008) <sup>7</sup>	Prince et al. (2010) <sup>13</sup>	Ganesan et al. (2015) <sup>14</sup>	Case 1*	Case 2*
Number of patients	7	6	5	1	1
Sex					
Male	6	3	5	0	1
Female	1	3	0	1	0
Diagnosis age in months	5.3	7.3	8	9	5
Average (range)	(0-11)	(5-10)	(2-12)		
Final age in years	6.2	3.2	4.4	1.3	3.4
Average (range)	(1.8-10.7)	(2- 5.6)	(4-5)		
Treatment	Observation	Observation	Observation (3)	Observation	Corset
			Corset (2)		
Initial angle	34.2°	32.8°	27.4°	18°	57°
Average (range)	(24° - 41°) sentado	(28°- 44°)	(15 -36°)	Standing	Sitting
Final angle	-0.4°	1.3°	7°	18°	19°
Average (range)	(-16 a 12°)	(0° a 5°)	(6 -8°)	Sitting	Sitting
Vertebrae involved	L1 and L2	L1 and L2	L2	L2	L2

subluxation and the clinical and radiological improvement as the bipedal position is reached<sup>8</sup>. It is recommended to reconsider the diagnosis before the clinical progression of the kyphosis, the absence of improvement when the gait is stabilized, and/or association with neurological or psychomotor developmental alterations and with the presence or appearance of other physical and/or skeletal anomalies. Publications from more than 50 years ago have described family cases, with inheritance consistent with an autosomal dominant pattern (OMIM 192900).

To date, a total of 18 cases of vertebral hypoplasia associated with kyphosis have been reported, all with similar characteristics to the cases described in this report (Table 1), regarding the age of diagnosis (range of 0 and 12 months), in relation to the vertebrae involved (L1 and L2), and to the favorable evolution <sup>13,14</sup>.

Probably due to the low prevalence of this disorder, there is still no consensus on its management. There are those who postulate only observation with periodic check-ups, while other authors advise the use of corsets to support the normalization of the spine<sup>14</sup>. The series with the highest number of cases is from Campos et al. who, in a retrospective review of 20 years, in a reference center in the United States, identified seven healthy children under three years of age with this diagnosis, who were observed clinically, without orthopedic support, showing a spontaneous resolution before six years of age8. In our patients, the first case had only observation, highlighting that once the march was established, clinical and radiological improvement was evidenced; while in the second case, orthopedic support was decided due to the significant degree of kyphosis at the time of diagnosis, superior to the others reported in the literature (Table 1), reaching a significant improvement at the age of three years.

#### **Conclusions**

In healthy infants, isolated vertebral hypoplasia associated with kyphosis may correspond to a minor abnormality or to an anatomical variant of favorable evolution. Clinical and radiological follow-up is recommended until normalization of lumbar curvature and vertebral defect.

#### **Ethical Responsibilities**

Human Beings and animals protection: Disclosure the authors state that the procedures were followed according to the Declaration of Helsinki and the World Medical Association regarding human experimentation developed for the medical community.

**Data confidentiality:** The authors state that they have followed the protocols of their Center and Local regulations on the publication of patient data.

Rights to privacy and informed consent: The authors have obtained the informed consent of the patients and/or subjects referred to in the article. This document is in the possession of the correspondence author.

#### **Financial Disclosure**

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#### **Conflicts of Interest**

Authors declare no conflict of interest regarding the present study.

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