Original Article

# Frequency and characteristics of congenital intraspinal abnormalities in a cohort of 128 patients with congenital scoliosis


## ABSTRACT

**Objectives:** The aims of this study were to determine the incidence and main characteristics of associated intraspinal anomalies in patients with congenital scoliosis (CS) and to analyze the different factors that influence the curve progression.

**Design:** This was a retrospective comparative study.

**Methods:** This was a retrospective study of 128 patients with CS.

**Main Outcome Measurements:** The incidence of the patients with intraspinal anomalies and their demographic, clinical, and radiological values was described.

**Results:** Intraspinal anomalies were present in 13.3% of the patients. Among them, the most frequent anomaly was syringomyelia. The most frequent curve was the thoracic curve. The main deformity based on McMaster classification was formation failure. The curve progression during follow-up did not show significant differences between vertebral anomalies, syringomyelia, presence of thoracic anomalies, and gender ($P > 0.05$).

**Conclusions:** Our study showed a lower percentage of spinal anomalies compared to other series. As other studies, the progression of the scoliosis curve in patients with spinal anomalies seems primarily to be determined by the type of vertebral malformation.

**Level of Evidence:** Level II.


**Keywords:** Abnormalities, congenital, intraspinal, scoliosis, spine

## INTRODUCTION

Congenital scoliosis (CS) is the presence of abnormal coronal plane curvature in the spine secondary to an anomalous congenital vertebral defect.[1] Large population studies utilizing screening low-dose radiography of the spine suggested a CS prevalence of 0.5%–0.11%.[2] The spine and spinal cord are closely related anatomically and developmentally; hence, it is not uncommon to see an intraspinal anomaly associated with CS.[3,4]

The prevalence of intraspinal anomaly associated with scoliosis has been reported between 15% and 47%.[1,3-16] It is crucial to diagnose intraspinal anomalies because they are usually hidden and can cause clinically important problems in CS.[13] In addition to the intraspinal anomalies, CS is related to


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other types of anomalies such as thoracic anomalies, cardiac anomalies, genitourinary anomalies, as well as a part of a complex syndrome or chromosomal abnormality.[15]

In patients with CS and intraspinal anomalies, the normal movement of the spinal cord in the spinal canal may be restricted, and any attempt to correct scoliosis could result in stretching of spinal cord and serious neurological complications. Therefore, it is necessary to detect intraspinal anomalies in these patients preoperatively.[6] Because of the rib cage associated with the thoracic spine, few studies have also analyzed the relationship between CS and rib anomalies.[15,17-19]

The primary goal of this study was to determine the incidence of associated intraspinal anomalies in our patients with CS and to analyze the type of intraspinal anomalies and associated pathology. The secondary goal was to evaluate the effect of these anomalies on the progression of the CS curve.

## METHODS

### Patients

A descriptive retrospective cohort study was performed. The institutional review board approval was obtained to retrospectively review all patients who had a diagnosis of CS between January 2013 and December 2017 at our institution (260 patients). Patients without complete computerized medical records (37 patients) and patients without an available magnetic resonance imaging (MRI) of the spine (95 patients) were excluded from the analysis. From the patients who met inclusion and exclusion criteria (128 patients), we identified the ones who had intraspinal anomalies associated with CS (17 patients).

### Data extraction

In these 17 cases, demographic data (age and sex), clinical data (diagnosis age of CS, follow-up, associated anomalies, and syndromes), and radiological data (type of deformity according to McMaster classification, initial Cobb angle at the time of diagnosis, curve apex location, and Cobb angle at the final follow-up) were collected. The site of the curvature was defined according to the classification proposed by the Scoliosis Research Society: cervical (apex between C2 and C6) cervicothoracic (apex at C7 or T1), thoracic (apex between T2 and T11), thoracolumbar (apex at T12 or L1), lumbar (apex between L2 and L4), and lumbosacral (apex at L5 or caudal).[20] All analyses and Cobb angles were measured by a single expert pediatric spine surgeon with the aim of eliminating interobserver variation. The first Cobb angle was the first radiograph and the last Cobb angle (postfollow-up) was the last radiograph. If the patient had been operated, we took the radiograph before surgery. We classified the patients according to the classification proposed by Winter *et al*.[21] and McMaster and Ohtsuka[22] into Group 1 – failures of vertebral formation, Group 2 – failures of vertebral segmentation, and Group 3 – mixed anomalies. To analyze the curve progression, only scoliotic curves were included. Patients with kyphosis of more than 20° were excluded. Curve progression was analyzed in relation to the vertebral defects as proposed by McMaster, their location, intraspinal anomalies, gender, and rib anomaly.

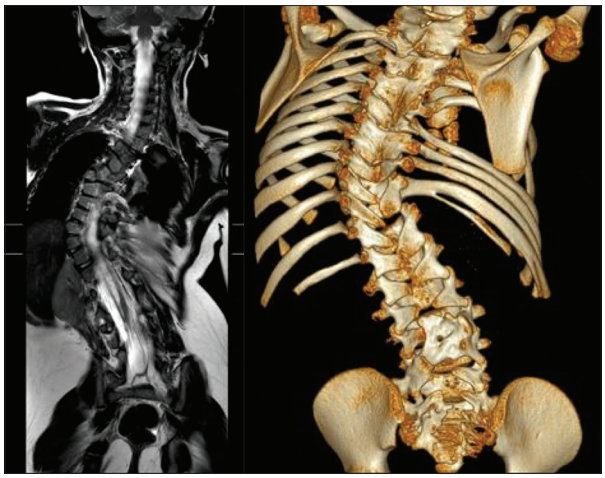

**Figure 1: A 13-year-old patient with syringomyelia, thoracic anomaly, and mixed defects**

### Statistical analysis

Statistical analysis was conducted using IBM SPSS version 24.0 (IBM Corp., Armonk, NY, USA). The categorical variables were described with their absolute values and percentages. The quantitative variables were presented by their measures of central tendency (mean and standard deviation). The incidence was calculated by dividing the number of patients with an abnormality by the total number of patients, and it was assessed as a cumulative measure and individually for each type of abnormality. The Chi-square test was used to evaluate the relationships between categorical variables. $P \leq 0.05$ was considered as statistically significant.

## RESULTS

### Demographic data

Between January 2013 and December 2017, a total of 128 patients met the inclusion and exclusion criteria (patients with CS with available MRI). Seventeen patients had intraspinal anomalies (13.3%). From these 17 cases, eight were men and nine were women, with a mean age of 10.1 ± 4.3. Other congenital anomalies were associated with intraspinal anomalies. There were eight patients with thoracic anomalies (47.1%), three patients had genitourinary anomalies (two – agenesis and one – horseshoe kidney), and there was one case of a cardiac anomaly (mitral valvulopathy).

### Intraspinal anomalies, location, deformity category, and syndromes

Table 1 summarizes these variables. The most prevalent intraspinal anomaly was syringomyelia in nine patients (52.9%) [Figure 1], followed by tethered cord (four patients). Two intraspinal anomalies were found in the same proportion: three patients with diastematomyelia and three patients with filum lipoma. In addition, dural ectasia, bifid spine, and arachnoid cysts were also seen. Some patients had more than one anomaly. The most frequent intraspinal anomaly associated with other spinal anomalies was tethered cord. The main curve locations were as follows: two curves were cervicothoracic, eight curves were thoracic, two thoracolumbar, and four lumbar. Only one patient had two main curves (one thoracic and other lumbar). According to the classification proposed by Winter and McMaster, ten patients had failures of vertebral formation: one was a butterfly vertebra, four totally segmented vertebra, one nonsegmented, and four patients had more than one formation defect; two patients had failures of vertebral segmentation and five patients mixed anomalies. The associated syndromes were Currarino syndrome, caudal regression syndrome, and Goldenhar syndrome. The syndromes were only observed in one patient each.

### Curve progression

Table 2 summarizes the different factors in relation to the curve progression. The average Cobb angle in all patients with intraspinal anomalies at initial diagnosis was 28.2 ± 15.7, and at the last follow-up (without surgery) (5.0 ± 3.7), it was 37 ± 26.6 (Dif = 9.1 ± 15.6; $P = 0.29$). According to McMaster classification, there was only one failure of segmentation, and therefore, only failures of formation and mixed failures could be compared. Patients with mixed failures showed progression of the Cobb angle by 17.3° ±16.62° and patients with formation

**Table 1: Demographic, clinical, and radiological information of the patients in our study**

| Serial number | Age | Gender | Main curve | Follow-up | Cobb pre | Cobb post | Cobb difference | Location | McMaster | Intraspinal anomalies | Location anomalies | Thoracic anomalies |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1 | 14 | Female | S | 10 | 29 | 42 | 13 | CT | Mixed | Syringomyelia | Cervical | - |
| 2 | 15 | Male | K | 13 | 15 | 31 | 16 | CT | Mixed | Syringomyelia | Dorsal | Fusion |
| 3 | 15 | Male | S | | | | | T and L | Totally segmented hemivertebra | Filum lipoma Tethered cord | Lumbar | - |
| 4 | 13 | Female | S | 4 | 35 | 25 | −10 | T | Multiple formation | Arachnoid cyst Tethered cord | Lumbosacral | Fusion |
| 5 | 9 | Male | S | 8 | 16 | 9 | −7 | TL | Totally Segmented hemivertebra | Filum lipoma | Lumbar | - |
| 6 | 10 | Female | K | | | | | L | Segmented | Lipomeningocele | Lumbar | - |
| 7 | 7 | Male | S | 6 | 62 | 75 | 13 | T | Mixed | Syringomyelia, diastematomyelia Spina bifida | Dorsal (T9–T10) Lumbar | Pectus carinatum |
| 8 | 14 | Female | S | 6 | 25 | 55 | 30 | T | Multiple formation | Chiari I | Cervical | Fusion |
| 9 | 16 | Male | S | 6 | 22 | 44 | 22 | T | Multiple formation | Syringomyelia | Dorsal (T4–T9) | Fusion |
| 10 | 7 | Female | S | 1 | 53 | 98 | 45 | T | Mixed | Diastematomyelia Syringomyelia | Dorsal (T5–T6) Dorsal (T3–T4) | - |
| 11 | 6 | Male | S | 5 | 8 | 5 | −3 | T | Totally segmented hemivertebra | Syringomyelia | Dorsal (T5–T11) | - |
| 12 | 4 | Female | S | 1 | 19 | 19 | 0 | T | Multiple formation | Diastematomyelia | Lumbar | - |
| 13 | 6 | Male | S | 1 | 23 | 20 | −3 | L | Semisegmented hemivertebra | Syringomyelia | Dorsal (T5–T12) | Fusion |
| 14 | 13 | Male | K | | | | | L | Totally Segmented hemivertebra | Dural ectasia | Lumbar | - |
| 15 | 6 | Female | S | 1 | 21 | 21 | 0 | TL | Mixed | Tethered cord, filum lipoma | Lumbar | Hypoplasia |
| 16 | 3 | Female | S | 2 | 18 | 17 | −1 | L | Butterfly | Syringomyelia, Tethered cord | Lumbar | - |
| 17 | 13 | Male | S | 3 | 47 | 56 | 9 | T | Segmented | Syringomyelia | Dorsal | - |

CT - Cervicothoracic; K - Kyphosis; L - Lumbar; S - Scoliosis; T - Thoracic; TL - Thoracolumbar

**Table 2: Valuation of the progression of the curve through different factors**

| | Cobb pre | Cobb post | Cobb difference |
|---|---|---|---|
| Formation failures ($n = 8$) | 21.0 ± 7.8 | 24.0 ± 17.1 | 4.0 ± 14.4 |
| Mixed failures ($n = 5$) | 36.2 ± 20.5 | 53.1 ± 32.2 | 17.0 ± 16.6 |
| *P* | 0.081 | 0.055 | 0.163 |
| Syringomyelia ($n = 9$) | 29.1 ± 18.9 | 42.4 ± 31.2 | 13.3 ± 16.13 |
| No syringomyelia ($n = 5$) | 23.2 ± 7.4 | 26.2 ± 17.6 | 3.0 ± 15.9 |
| *P* | 0.514 | 0.315 | 0.286 |
| Thoracic + intraspinal ($n = 7$) | 29.3 ± 15.7 | 39.3 ± 20.5 | 10.0 ± 14.5 |
| Intraspinal ($n = 7$) | 27.1 ± 16.9 | 35.3 ± 33.2 | 8.2 ± 17.7 |
| *P* | 0.823 | 0.791 | 0.821 |
| Male ($n = 7$) | 23.0 ± 17.7 | 29.3 ± 24.3 | 6.3 ± 11.1 |
| Female ($n = 7$) | 33.3 ± 12.8 | 45.4 ± 28.1 | 12.1 ± 19.5 |
| *P* | 0.249 | 0.276 | 0.492 |

*P*: $P < 0.005$

failures progressed by 4.0° ±14.4° during follow-up ($P = 0.16$). The most frequent intraspinal anomaly (syringomyelia) was compared with the other anomalies, and there was no significant difference between the two groups: Cobb angle 13.0 ± 16.1 versus 3.3 ± 15.9 ($P = 0.286$). Thoracic anomalies associated with intraspinal anomalies did not show any difference in curve progression than the intraspinal anomalies alone (10 ± 14.48 vs. 8 ± 17.63; $P = 0.821$). Finally, for females, progression of the curve was measured 12° ±19.48° vs. 6° ±11.10° in males, and this was without significant difference ($P = 0.492$).

## DISCUSSION

The vertebral column and spinal cord are closely related anatomically and developmentally. The most important finding of this study was that our study showed a lower percentage of spinal anomalies compared to other series (13.3%). Syringomyelia was the most frequent intraspinal anomaly. The definition of syringomyelia was the presence of a fluid cyst within the spinal cord. The presence of a prominent central canal was not considered syringomyelia.

In our study, the prevalence of intraspinal anomaly associated with CS was 13.3%. This value differs from previous reports, which report a higher percentage of intraspinal anomalies (15%–47%).[1,3-16] McMaster[3] reported intraspinal anomaly in 18% of 251 patients with myelography. Using MRI as in our study, Bradford *et al*.[10] reported 38% of intraspinal anomaly in 42 United States patients with CS. Liu *et al*.[1] evaluated 539 Chinese patients and found intraspinal anomalies of 24.5%. Gupta *et al*.[6] reported 47% of intraspinal anomaly in 119 Indian patients. We think that the lower prevalence found could be related to different populations in different series. To our knowledge, this is the first study which evaluated intraspinal anomalies associated with CS in Spain. Bollini *et al*.[16] evaluated 75 French patients and found intraspinal anomalies of 15%, similar to our study. It has been proposed that oxygen concentration in different geographic regions (such as high altitude) could also be a risk factor.[9,18,23] In animals when the pregnant female mice were exposed to hypoxia, most of the offspring developed congenital spinal deformities, including fused, split, or hemivertebral bodies and rib abnormalities.[24] Furthermore, Hou *et al*.[9] found that in China, CS patients in high-altitude geographic regions tend to have a higher proportion of intraspinal malformations than patients with CS in low-altitude geographic regions. In Bollini *et al*.[16] and in our study, both cities are at sea level altitude, Marseille, and Barcelona, respectively.

In patients with CS and intraspinal anomalies, the normal movement of the spinal cord in the spinal canal may be restricted.[6] This may affect the supporting musculature of the spine and may influence the scoliosis progression. In this study, the total progression of these patients was 9°, with an average follow-up of 5 years. The literature suggests that rib anomalies may adversely affect the progression of the spinal deformity. Shahcheraghi and Hobbi in their study of sixty patients with CS, of which 16 patients had fusions of the ribs, found that fused ribs on the concave side of a lower thoracic curve increased the rate of curve progression.[25] Tsirikos and McMaster study showed that congenital rib anomalies occur most frequently on the concavity and either simple or complex rib abnormalities do not appear to have an adverse effect on the size or the rate of progression of the curve.[26] In this study, we found a high number of rib anomalies associated with intraspinal anomalies. In our opinion, we think that rib fusions on the concavity of a scoliosis can contribute to the development of the curvature. However, our study primarily assessed patients with CS and intraspinal anomalies.

With regard to the type of anomaly, the highest progression potential was related to CS due to mixed defects but without a statistically significant difference. However, it is important to take into account what is statistically significant and what is radiologically significant. Patients with mixed defects show a curve progression of 17° compared with the formation of defects (4°). An additional 13° of curve progression may change management in clinical practice. In the formation failure group, there was a high variety of defects, and it was observed that the totally segmented hemivertebras, semisegmented vertebras, and butterfly vertebras did not progress. Nevertheless, patients with more than one formation defect progressed by 11°. This confirms that mixed defects are the most likely to progress, as has been shown before.[18,19,27]

Patients with clinical syndromes tend to have more than one anomaly, and involvement of other systems is common. This

can significantly impact the patients' overall management strategy. Therefore, a careful and comprehensive preoperative evaluation and discussion is required, before considering any surgical management.

One of the limitations of this study is the retrospective nature of our study. A second limitation is that this is a single institution study with a low incidence of intraspinal anomalies. Therefore, subgroup analysis and comparisons between different intraspinal defects and curve progression could not be performed.

However, our institution is a reference hospital of this pathology, and we were able to analyze 128 MRI to assess the incidence and characteristics of intraspinal anomalies in CS patients. This is a fairly large number of patients with MRI evaluation of intraspinal anomalies, as compared to available literature.[1,3-16] Despite the limitations, this is the first study which evaluated intraspinal anomaly associated with CS in Spain.

## CONCLUSIONS

In summary, this study details the characteristics of the congenital intraspinal anomalies in a large cohort of CS. The different factors that affect the progression of the curve should continue to be investigated, although the progression of the curve in patients with spinal anomalies seems to be determined to a greater extent by the type of vertebral malformation.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

### Conflicts of interest

There are no conflicts of interest.

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